

Version 9 **User Guide** 

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#### Preface

Welcome to the Progeny Software User's Manual. The purpose of the Progeny Software User's Manual is to answer your questions and guide you through the procedures necessary to use the Progeny Clinical application, the Progeny Lab application, and the Progeny LIMS application efficiently and effectively.

# Using the manual

You will find the Progeny Software User's Manual easy to use. You can simply look up the topic that you need in the table of contents or the index. In this Preface, you will find a brief discussion of each chapter to further assist you in locating the information that you need.

# Special information about the manual

The Progeny Software User's Manual has a dual-purpose design. It can be distributed electronically and then printed on an as-needed basis, or it can be viewed online in its fully interactive capacity. If you print the document, for best results, it is recommended that you print it on a duplex printer; however, single-sided printing will also work. If you view the document online, a standard set of bookmarks appears in a frame on the left side of the document window for navigation through the document. For better viewing, decrease the size of the bookmark frame and use the magnification box to increase the magnification of the document to your viewing preference.



If you do print the document using a single-sided printer, you might see a single blank page at the end of some chapters. This blank page has been added solely to ensure that the next chapter begins on an odd-numbered page. This blank page in no way indicates that your book is missing information.

### Conventions used in the manual

The Progeny Software User's Manual uses the following conventions:

- Information that can vary in a command—variable information—is indicated by alphanumeric characters enclosed in angle brackets; for example, <Sample ID>. Do not type the angle brackets when you specify the variable information.
- A new term, or term that must be emphasized for clarity of procedures, is italicized.
- Page numbering is "online friendly." Pages are numbered from 1 to x, starting with the cover and ending on the last page of the index.
- The Progeny application provides multiple ways of accessing the same Progeny functions. If an option is available from the toolbar as well as other areas (for example, the main menu or a context menu), then this manual references the toolbar. Although this manual does not address all the multiple ways of accessing the same function, you can use whatever method best suits your working needs.
- This manual is intended for both print and online viewing.
- If information appears in blue, it is a hyperlink. Table of Contents and Index entries are also hyperlinks. Click the hyperlink to advance to the referenced information.

# Assumptions for the manual

The Progeny Software User's Manual assumes that you are familiar with Windows-based applications and basic Windows functions, navigational elements, and icons.

## Organization of the manual

In addition to this Preface, the Progeny Software User's Manual contains the following sections and appendices:

- Section 1, "Progeny Core Database Functionality" details the necessary steps for designing and maintaining a
  Progeny database that is necessary for data storage, data retrieval, and ensuring the integrity of the data in
  your database.
- Section 2, "Progeny Clinical" details the complete functionality of the Progeny Clinical module, which is pedigree and clinical data management software that you use for family-based studies.
- Section 3, "Progeny Lab" details the complete functionality of the Progeny Lab module, which is software that is designed for the management of whole genome association, targeted genotyping, and linkage studies.
- Section 4, "Progeny LIMS" details the complete functionality of the Progeny LIMS module, which is software that is designed for managing any type of sample and associated data in a fully customizable multi-level inventory system with the freedom to create any laboratory workflow.
- Chapter A, "System Fields" details the read-only fields that Progeny automatically stores for a record. Each record type (individual, pedigree, sample, and marker) has its own unique system fields.
- Chapter B, "Importing Clinical Data" details the Import module to import external pedigrees, individuals, and samples that do not already exist in a Progeny database. You can also use the Import module to import data that updates individuals and samples that already exist in the database.

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# Section 1 – Progeny Core Database Functionality

#### This section contains the following chapters:

- Getting Started with Progeny
- Progeny Database Design
- Datasheet Basics
- Spreadsheet Basics
- Database Security
- Database Reporting
- <u>Database Organization</u>
- Database Queries and Searches
- Database Auditing
- Database Backup

# Chapter 1 – Getting Started with Progeny

Progeny software is a suite of three individual products—Progeny Clinical, Progeny Lab, and Progeny LIMS — with each product designed for data management in a specific area. Progeny Clinical is designed for the management of clinical data, Progeny Lab is designed for the management of genotype data, and Progeny LIMS is designed for the management of sample data and inventories. Progeny Clinical, Progeny Lab, or Progeny LIMS can be combined in any fashion (including all three products) into a single application that is ideal for complete family history data management. This chapter provides an overview of the Progeny main window and its major navigational elements.

This chapter covers the following topics:

• Launching Progeny and the Progeny Main Window

## Launching Progeny and the Progeny Main Window

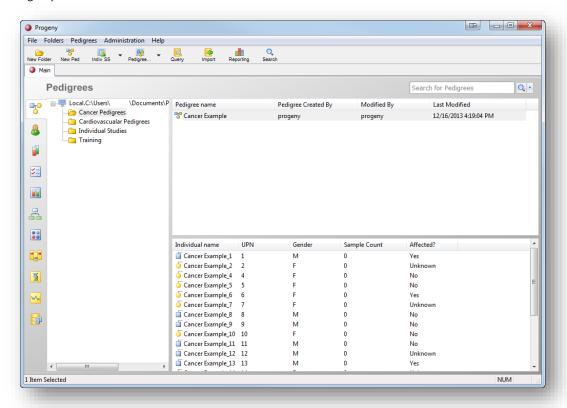
After your administrator installs and registers Progeny, a shortcut icon for the application is placed on your desktop. An option for the application is also available from your Start menu. You can double-click the desktop icon, or you can select the option from your Start menu to open the Login window.

Figure 1-1: Progeny desktop icon



After you log into the application, the Progeny main window opens. The Progeny main window is your starting point for the Progeny application. The Progeny main window has six major components—the <u>panes</u>, the <u>title bar</u>, the <u>main menu</u>, the <u>navigation bar</u>, the <u>toolbar</u>, and the <u>tab bar</u>.

Figure 1-2: Progeny main window



#### **Panes**

The Progeny main window has three panes. The left vertical pane displays the folder organization for a functional area in classic Windows Explorer-style structures. Click on a folder in the pane to open the folder. As folders and sub-folders are opened in this pane, detailed information about the folder's content is displayed in the upper and lower right horizontal right panes. (Information is displayed in the bottom pane when an item is selected above.) Right-click menus are available for all folders and list items which are context specific to not only the pane but also the item type.



The folders displayed in the upper-left pane will correspond to the module you have selected (pedigrees, individuals or samples, for instance). The panes to the right correspond to parent items on top and sub-items below. For example, if you are in the Pedigrees module and select a pedigree, all the individuals attached to that pedigree will be displayed in the lower-right pane. Likewise, if you are in the Individuals module and select an individual then all samples attached to that individual will be displayed in the lower-right pane.

Figure 1-3: Progeny main window panes

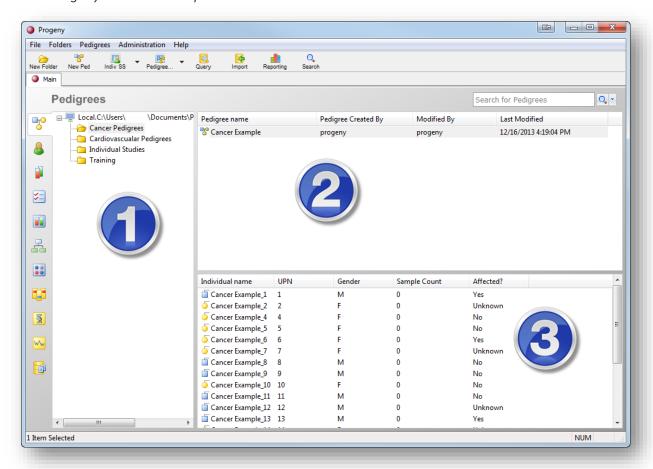
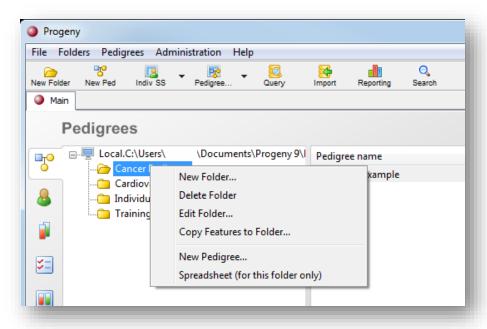


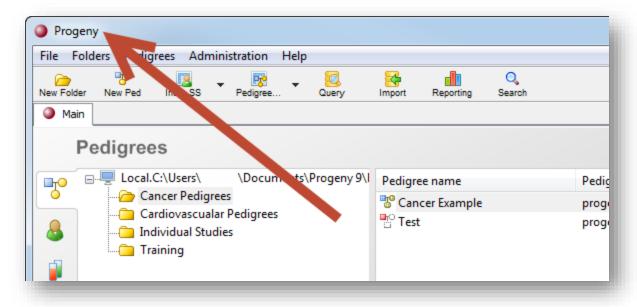
Figure 1-4: Example of a folder context menu



### Title bar

The name "Progeny" appears in the title bar at the top of the Progeny main window. (The version number is not displayed in the title bar. The version number can be found by selecting Help  $\rightarrow$ About Progeny from the Main Menu.)

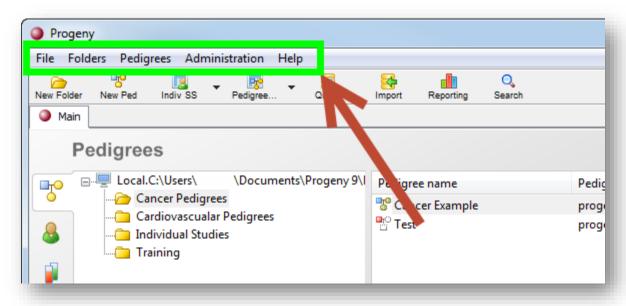
Figure 1-5: Title bar



#### Main menu

The main menu is set up in a standard Windows menu format with menu commands grouped into basic categories (File, Folders, Pedigrees, Administration and Help) across the menu bar. Many of the menu options are context related and will vary depending on which tab you have selected in the Navigation bar. Other options are reserved for administrative accounts such as the 'progeny' (admin) user account.

Figure 1-6: Main menu



#### Navigation bar

The navigation bar is the vertical bar displayed on the left side of the main window. The navigation buttons take you through the main screens of each of the three products of the Progeny Suite (Clinical, Lab, and LIMS). Three of the navigation buttons (Samples, Tasks, and Fields) are common to all three products, while the others are product specific.



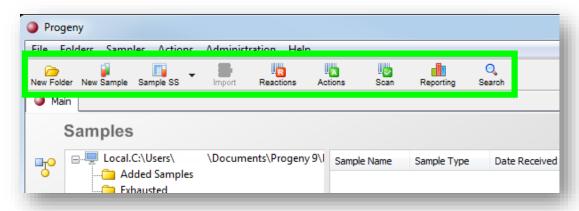
When Progeny is installed, the navigation buttons are displayed in a default order on the navigation bar. You can change the order of the buttons on the Navigator bar to better suit your working needs simply by dragging a button to a new location.

Product	Button	Description
Common	<b>\$</b> =	Tasks—The Tasks feature centralizes any task that has been flagged with a reminder in the database in a single location. When you log into Progeny, if any tasks have been flagged with a reminder, then the Tasks main window opens with these tasks displayed on a peruser basis. The Tasks button is displayed for all three Progeny products.
	<b>1</b>	Fields button—The Fields button opens the Fields main window. The window has options for creating, modifying and deleting data folders and fields in the database. The Fields button is displayed for all three Progeny products.
		Samples button—The Samples button opens the Samples main window. The window has options for creating, organizing and storing data sample information in the database. The Samples button is displayed for all three Products (although more capabilities are available in this section with Progeny LIMS).
Clinical	3	Pedigrees—The Pedigrees button opens the Pedigrees main window. The window has options for creating, organizing and storing pedigrees in the database. The Pedigrees button is displayed only if your organization has purchased and installed Progeny Clinical.
	8	Individuals—The Individuals button opens the Individuals main window. The window has options for creating, organizing, and storing individual records in the database. An individual, can be a singlet or be a member of a pedigree. The Individuals button is displayed only if your organization has purchased and installed Progeny Clinical.
Lab		Markers button—The Markers button opens the Markers main window. The window has options for creating, organizing and storing marker maps and associated marker information in the database. The Markers button is displayed only if your organization has purchased and installed Progeny Lab.
	5	Genotypes button—The Genotypes button opens the Genotypes main window. The window has options for importing and managing genotype data in the database. The Genotypes button is displayed only if your organization has purchased and installed Progeny Lab.
		Ww
		Inventory button—The Inventory button opens the Inventory main window. The window includes a graphical interface for complete multi-level inventory and individual and batch sample tracking and auditing. The Inventory button is displayed only if your organization has purchased and installed Progeny LIMS.
TIMS		Workflows button—The Workflows button opens the Workflows main window. The window has options for creating, organizing and storing sample workflows in the database. The Workflows button is displayed only if your organization has purchased and installed Progeny LIMS.
	• •	Containers button—The Containers button opens the Containers main window. The window has options for creating, organizing and storing container information in the database. The Containers button is displayed only if your organization has purchased and installed Progeny LIMS.

#### Toolbar

The toolbar is located just below the main menu. The toolbar provides quick access to all the available functions for a selected product feature. The toolbar is dynamically updated based on the window that is open.

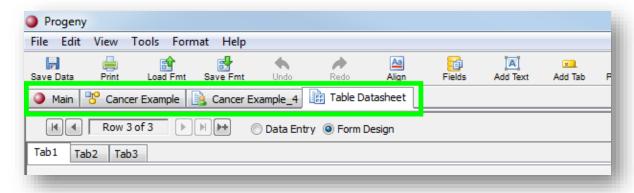
Figure 1-7: Example of the toolbar when Samples is selected



### Tab bar

Datasheets, spreadsheets, lookup tables, tables, and pedigrees open as separate tabs just below the toolbar in the main window. To open a datasheet, spreadsheet, table, or pedigree, simply click the tab for it. You can return to the main window at any time by clicking the Main tab. To close a tab, right-click on the tab and select Close Tab or click the red "X" at the far right of the tab.

Figure 1-8: Tab bar



# Chapter 2 – Progeny Database Design

At a very high level, database design is the logical design of the base data structures that are used to store the data in the database. Complete and accurate database design is critical for data storage, data retrieval, and ensuring the integrity of the data in your database. This chapter details the procedures for creating, maintaining, and modifying a Progeny database.

This chapter covers the following topics:

- Creating a Progeny Database
- Progeny Record-Level Database Fields
- Database Field Types
- Adding, Editing, and Deleting Database Fields
- Adding a Table
- Adding a Lookup Table
- Adding a Computed Field
- Adding a Summary Field
- Adding a Table Summary Field
- Copying Fields between Databases
- Setting Validations for a Database Field
- Adding, Editing and Deleting Database Folders

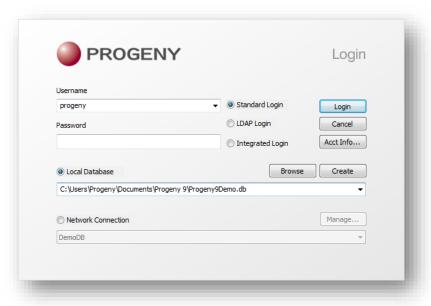
### **Creating a Progeny Database**

When the Progeny desktop client is installed, the demo database (Progeny9Demo.db) is installed to the Documents\Progeny 9 folder. This database contains basic fields and formats for creating cancer or cardiovascularrelated pedigrees. There is also a folder containing a training pedigree which will provide simple steps for building a pedigree and completing the Individual Datasheet. You may modify this database as needed to create your own Progeny database, use it as a template for creating other Progeny databases or merely draw inspiration from it to create your own Progeny database completely "from scratch." Progeny databases have unique structuring and as such can only be opened within a Progeny application. Additionally, the Progeny application can only open databases that were created by a Progeny application. You are free to create as many databases as needed to support your business needs.

#### To create a Progeny database

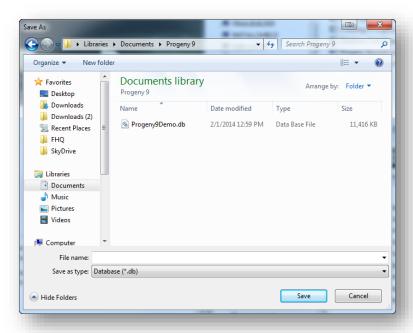
1. Launch the Progeny application.

Figure 2-0: Progeny launch screen



- 2. On the Login window, click Create.
  - a. The Save As dialog box opens. The Save as Type field is automatically populated with the file type of Database (.db) and you cannot change this value.

Figure 2-1: Save As dialog box

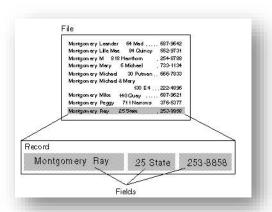


- 3. Browse to the location in which the database is to be saved, and then in the File name field, enter a name for the database.
- 4. Click Save.
  - a. You can now add fields to the new database and/or copy fields from the sample database or another database into the new database. See <a href="Adding, Editing, and Deleting Database Fields">Adding, Editing, and Deleting Database Fields</a> and Copying Fields between Databases.

# **Progeny Record-Level Database Fields**

A database field is a single piece of information from a record. A database record is a set of fields. A file is a collection of records.

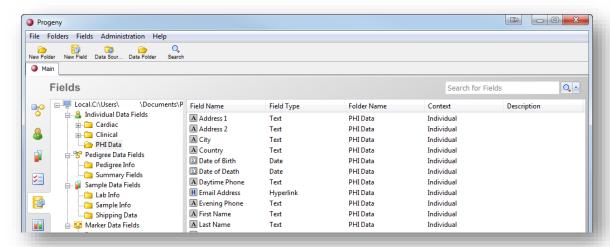
Figure 2-2: Example of the components of a database



The Fields window displays the record-level fields that are contained in a Progeny database. To open this window, click the Fields button on the Navigation bar. There are four different types of record-level fields in Progeny:

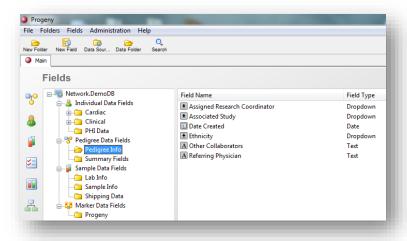
1. Individual database fields – Individual database fields are stored at the individual record level. The demo database that is shipped with Progeny contains individual data fields grouped into Cardiac, Clinical and PHI Data folders with additional subfolders under Cardiac and Clinical. Individual database fields are displayed on the Fields window only if your organization has purchased and installed Progeny Clinical.

Figure 2-3: Individual database fields in Progeny demo database



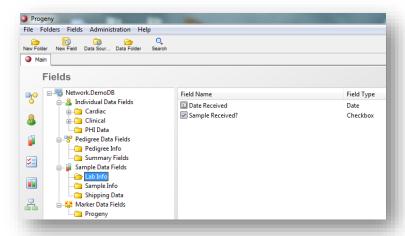
Pedigree database fields – Pedigree database fields are stored at the pedigree record level. The demo
database that is shipped with Progeny contains pedigree data fields grouped into Pedigree Info and
Summary Fields folders. Pedigree database fields are displayed on the Fields window only if your
organization has purchased and installed Progeny Clinical.

Figure 2-4: Pedigree database fields in Progeny demo database



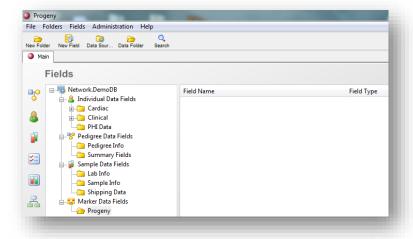
3. <u>Sample database fields</u> – Sample database fields are stored at the sample record level. The demo database that is shipped with Progeny contains sample data fields grouped into Lab Info, Sample Info and Shipping Data folders. Sample database fields are displayed with all products of the Progeny Suite.

Figure 2-5: Sample database fields in Progeny demo database



<u>Marker database fields</u> – Marker database fields are stored at the marker record level. The demo database that is shipped with Progeny contains a progeny folder but does not include any fields. Marker database fields are displayed on the Fields window only if your organization has purchased and installed Progeny Lab.

Figure 2-6: Marker database fields in Progeny demo database



# **Database Field Types**

When creating database fields in a Progeny database, you must specify the type for the field. The field type determines the type of data that a user can enter into the field (alphanumeric, numeric, date, and so on) or determines how the field functions in the database, for example, an image displays in the field when a user selects an image file for an image field, dropdown fields provide a custom picklist and date fields provide a date and time picker when end users are doing data entry for those field types. By default, each field type is marked with a unique icon in the application. Not all field types are available for all record levels. Table 2-1 below lists the field types that are available for each record level field in a Progeny database. It also provides a description of each field and the records for which the field type is applicable.

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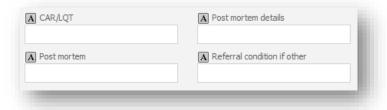
Table 2-1: Progeny database field types

Field Type	Icon	Description	Individual	Pedigree	Sample	Marker
Text	A	Alphanumeric characters (all digits and printable characters) up to a maximum of 32,000 characters.		Х	х	х
Date	2	M/D/YY or M/D/YYYY X			Х	х
Numeric	8	The digits 0-9. Negative numbers and decimals allowed. X			х	х
Yes/No	V	Checkbox field that holds a value of either 1 (to indicate Yes) or 0 (to indicate No).		х	Х	Х
Dropdown	ŧ	Displays a list of pre-defined list of items from which the user can select one item.			Х	Х
Image		Allows for the display of an image in an allowed format in the field.	х	Х	Х	Х
Table	EH .	The field is to be included in a table. The field does not have a pre-defined value.	х	х	Х	Х
Lookup Table		The field is to be included in a table. The fields have predefined values.		х	Х	
Computed	乜	No data entry is allowed in this field. Instead, a non-editable value is displayed in the field. The value that is displayed is automatically calculated based on the expression, function, or constant assigned to the field or values that are contained in other fields.	yed in the field. The value that is displayed is calculated based on the expression, function, or X gned to the field or values that are contained in		х	х
Summary	Σ	Provides summary information for a given column on a per pedigree basis. The summary can be an average, a total count, a minimum value, a maximum value, or a sum.	х	х	Х	
Table Summary	<b>3</b>	A computed field that summarizes the data for a given column in a given table.	х	х	Х	Х
Hyperlink	H	Links to an external document, website, and so on.	х	х	Х	Х

#### Text field

A text field is for alphanumeric data (all digits and printable characters). You can enter up to a maximum of 32,000 characters in a text field. A text field is the most generic and least restrictive of all the available field types. Examples of text fields include First Name, Last Name, Address, City, Notes and so on.

Figure 2-7: Text field example



#### Date field

A date field contains a calendar. You can use the calendar to enter a date in a date field, or you can simply enter the date in the field. The first time that you click on a date field, the checkbox for the field is not selected, which means that the field is not editable. To make the field editable, you can do one of the following:

- Press the spacebar on your keyboard.
- Select the checkbox in the date field.
- Use the calendar by clicking on the drop-down arrow in the field, and then scrolling to and clicking on the appropriate date.

A date is stored in the database in a four-digit format (M/D/YY) regardless of the date's display when you use the calendar (MM/DD/YYYY) or if you manually entered a four-digit year. Dates are displayed in a date field based on the regional settings for the client.

Figure 2-8: Date field example



#### Numeric field

You can enter only the digits 0 through 9 in a numeric field. Decimals and negative numbers are allowed; however, no other characters, either alphabetic or special (such as dashes or commas) are allowed. If you want to sort data based on numeric values, then you must use a numeric field. You cannot sort based on numeric values if the data is entered in a text field.

Figure 2-9: Numeric field example



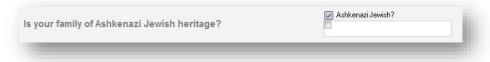
### Yes/No field

A Yes/No field is a checkbox field that holds a value of either 1 (to indicate Yes) or 0 (to indicate No). Progeny always stores a value in a database for a checkbox field whether the field is checked. If the field is checked, Progeny stores a value of "1" in the database. If the field is not checked, Progeny stores a value of "0" in the database.



For some legacy Progeny databases (Progeny 6 and earlier), if a checkbox field has never been edited (meaning selected and then cleared) it registers as Null, not No. You must select the checkbox and then clear it for it to register as No or update these values within the spreadsheet. You should use the Yes/No field for fields where you want to track only Yes values. To track both Yes and No values, you should use a Dropdown field with the values "Yes" and "No". All Y/N fields created in Progeny 7 or later will generate default values of "N" rather than Null.

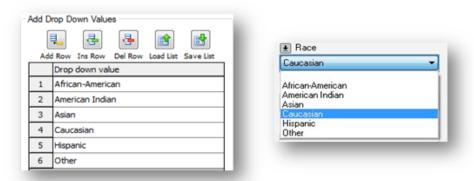
Figure 2-10: Yes/No field example



### Dropdown field

A drop-down field displays a pre-defined list of items from which you can select. To open the list, simply click in the field. You can scroll through the list and select one item at a time. The items are mutually exclusive. For example, you might create a dropdown field named "Race," and the available items are African American, American Indian, Asian, and so on. When you are adding/editing a drop-down field, the order in which you enter the items on the Add New Field/Edit Field dialog box is the order in which the items are displayed in the dropdown list. You can use the arrow keys on your keyboard to move up and down the list of available values for the field. If you are selecting from a lengthy dropdown list on a datasheet, you can enter the first few letters of the name of the item that you want to select, and the list is dynamically updated with the items that meet your search criteria.

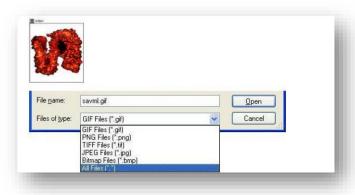
Figure 2-11: Add Dropdown Values pane on the Add New Field/Edit Field dialog box and corresponding Dropdown field



# Image field

An image field can display an image file in one of the following formats—.bmp, .gif, .png, .tiff, and .jpg/.jpeg. When you click on an image field, you are prompted to enter the type of image file that you want to display. You then scroll to the appropriate image file and select it. After you select the image file, the image is displayed in the Image field.

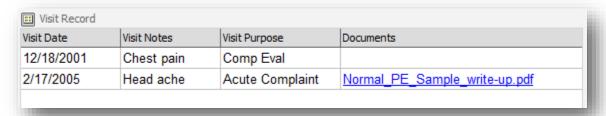
Figure 2-12: Image field example



#### Table

A table allows you to enter multiple values for the same set of data fields. For example, you might add a table called Visit Information that would allow a user to enter Visit Date field (a date field), the Visit Type field (a text field), and the Visit Purpose field (a drop-down field)

Figure 2-13: Table field example



# Lookup Table

A lookup table contains a dropdown list of fields that are related in some way. Each row within the drop-down contains a pre-defined "lookup" value which refers to a set of data fields that can be updated to remain current. For example, you might create a lookup table called Referring Physician that would allow a user to simply select a physician's name from the list, thereby associating all the physician's current contact information such as the physician's phone number, the physician's pager number, institution, and ID. To enter data in a lookup table field, simply click in the field to open the table, and select the appropriate value from the drop-down table.

Figure 2-14: Lookup table field example



### Computed field

You cannot enter data into a computed field. Instead, a computed field contains an expression that yields a value that is automatically calculated each time the window that contains the field is opened. For example, a computed field for Current Age is defined as the difference in years between the date that is in the Current Date field and the date that is in the Date of Birth field.

Figure 2-15: Computed field example



### Summary field

The purpose of a summary field is to summarize the data of object sub items (individuals or samples) of a database object (a pedigree, individual or sample). A summary field provides summary information for any selected data field and displays a summary value as a maximum value, minimum value, average, sum or count. Summary functions can be carried out on all fields that are not contained in tables and conditional queries can be employed to populate precise summary values. Pedigree level summary fields summarize individual-level data of the individuals within each pedigree. Individual-level summary fields summarize sample level data of the samples associated with everyone. And sample level summary fields summarize sample level data of the sub-samples and aliquots of each sample. For example, a pedigree level summary field might provide a count for the total number of cancer diagnoses in the pedigree. An individual level might provide a count of the number of available DNA samples. And a sample level summary might provide a total available volume of all non-exhausted aliquots of each sample.

Figure 2-16: Summary field example



### Table Summary field

Like a summary field, a table summary field is a computed field that summarizes a given column of a table. For example, you might create a table called Visit Information with a field in it called Visit Date and you want to see what the most recent visit date is for a specific patient. You can create a table summary field called Most Recent Visit Date that displays the most recent visit date for a patient that has five distinct visit date entries in the Visit Record table.

Figure 2-17: Table summary field example



### Hyperlink field

A hyperlink field contains a hyperlink to an external file, program or website. Simply click on the link in the field to open the file, program, or website. For example, a hyperlink field named Email could contain an individual's email address. When you click on the email address in the field, an untitled email addressed to the individual opens in the available email client such as Microsoft Outlook. Hyperlink fields can also be used to load and view documents.

Figure 2-18: Example of a hyperlink field



# Adding, Editing, and Deleting Database Fields

You can add, edit, and delete record-level database fields only from the Fields window. When adding recordlevel fields in a Progeny database, you must specify the field type. You can also set a default value for the field, add validation rules to the field, and set security for the field. You can also create folders for storing and organizing database fields. You can also delete a database field. When you delete a database field, it deletes not only the field but also, any data that is stored in the field.



The remainder of this procedure details the high-level steps for adding new database fields to a database, regardless of the data type. If you simply want to copy existing fields from another database into a new database, see Copying Fields between Databases

#### To add a database field

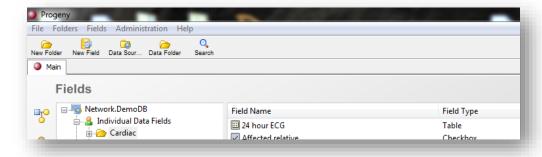


Make sure that you are logged in to the database to which you want to add the fields.



- 1. On the Progeny main window navigation bar, click the Fields button
  - a. The Fields window opens.

Figure 2-19: Fields window

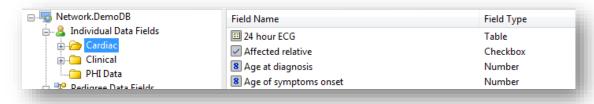


- 2. Determine the record type for which you are creating the field (Individual, Pedigree, Sample, or Marker). In the left pane of the window, open the database folder in which you are adding the field.
  - a. A list of the fields that are already contained in the folder is displayed in the right pane of the Fields window.



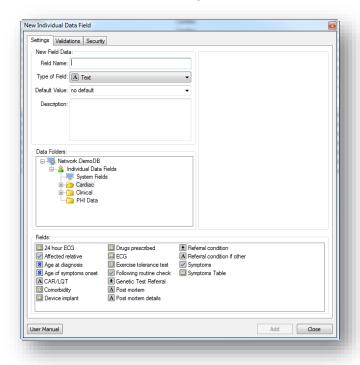
For information about organizing database folders in folders, including adding new folders, see <u>Adding</u>, <u>Editing and Deleting Database Folders</u>

Figure 2-20: Selected records folder with fields displayed in the Fields window



- 3. On the Progeny toolbar, click the New Field button New Field
  - a. The New Data Field dialog box opens. The Settings tab is the active tab for the dialog box. All the fields that are contained in the selected folder are displayed alphabetically in the lower pane of the dialog box.

Figure 2-21: Example of a New Individual Data Field dialog box



- 4. In the Field Name dialog box, enter the name for the new field.
- 5. On the Type of Field dropdown list, select the type for the new field.
  - a. The field type determines the type of data that a user can enter into the field or it determines how the field functions in the database. Not all field types are available for all record types. (See <u>Database Field Types</u>) In addition, if you are adding any of the following field types—Table, Lookup Table, Computed, Summary, or Table Summary field—then you must carry out additional steps to add the database field. For additional information refer to:
    - i. Adding a Table
    - ii. Adding a Lookup Table
    - iii. Adding a Computed Field
    - iv. Adding a Summary Field
    - v. Adding a Table Summary Field
- 6. Optionally, do one or more of the following:
  - a. If available, in the Default Value field, enter the default value for the field. A user can always edit this value.
  - b. Open the Validations tab and set the validation rules for the field. See <u>Setting Validations for a Database Field</u>
  - c. Open the Security tab and set the security for the field. See Setting Database Field Security
- 7. Click Add. A message opens indicating that the new field was added successfully.
- 8. Click OK. The message closes. The Add New Field dialog box remains open. The new field is displayed in the lower pane of the dialog box.

#### To edit a database field

- 1. On the navigation bar, click the Fields button . The Fields window opens.
- 2. In the left pane of the window, select the folder that contains the field that you are editing.
  - a. A list of the fields that are contained in the folder is displayed in the right pane of the Fields window.
- 3. Double-click the field that you are deleting.
  - a. The Edit Field dialog box opens. The dialog box displays all the current information for the field.
- 4. Edit the information for the field, including the field name, as needed. For additional information refer to:
  - a. To modify the validation settings, see Setting Validations for a Database Field
  - b. To modify the security settings, see Setting Database Field Security
- 5. Click Change.
  - a. A message opens asking you if you are sure that you want to change the field.
    - i. Click Yes. The message and Edit Field dialog box close. You return to the Fields window with the fields listed in the right pane.

#### To delete a database field

You can delete a database field only if no datasheets, spreadsheets, or pedigrees are open. If any of these items are open, right-click on the item's tab and click Close Tab. You can then continue with deleting database fields.

1. On the navigation bar, click the Fields button



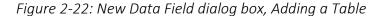
- a. The Fields window opens.
- 2. In the left pane of the window, on the Main tab, select the folder that contains the field that you are deleting.
  - a. A list of the fields that are contained in the folder is displayed in the right pane of the Fields window.
- 3. Right-click on the field that you are deleting (CTRL-click to select multiple fields), and on the context menu that opens, click Delete Field.
  - a. A message opens asking you if you are sure that you want to delete the selected fields and that the operation will delete the fields and the data.
- 4. Click OK. The message closes and the field (and the data stored in the field) is deleted from the database.

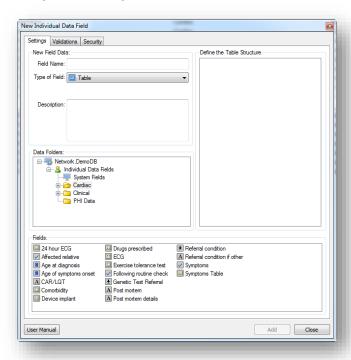
# Adding a Table

Before you add a table, you must confirm that the fields that are to be added to the table already exist in the database, otherwise, you must add them. For example, before adding a table called Visit Information, you need to confirm that the fields Visit Date, Visit Type, and Visit Purpose already exist in the database (see <a href="Contextual Search">Contextual Search</a>), otherwise, you must add them.

#### To add a table

1. On the New Field dialog box, enter the name for the Table field and select Table for the field type.



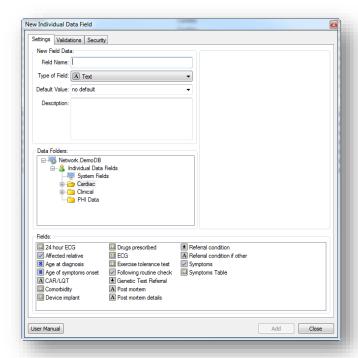


- 2. In the Data Folders pane, open the data folder that contains the fields that are to be added to the Table.
  - a. The fields that are contained in the data folder are displayed in the Fields pane at the bottom of the Add New Data Field dialog box.
- 3. Drag each field that you are adding to the Table from the Fields pane to the Define the Table Structure pane (upper right pane) of the Add New Data Field dialog box.
- 4. In the Data Folders pane, select the folder where you would like to save your table.



The order in which the fields are displayed in the Define the Table Structure pane is the order in which the rows are displayed in the table. You can change this order if needed. See <u>To format a table in a datasheet</u>

Figure 2-23: New Data Field dialog box, Defining the Table structure



- 5. Click Add. A message opens indicating that the new field was added successfully.
- 6. Click OK. The message closes.
- 7. Click Close in the Add New Field dialog box to close the dialog box and return to the Fields window.

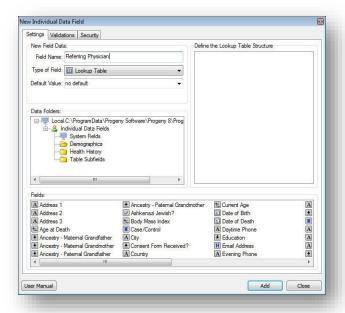
# Adding a Lookup Table

Before you add a lookup table, you must confirm that the fields that are to be added to the lookup table already exist in the database, otherwise, you must add them to the database. For example, before adding a lookup table called Referring Physician, you need to confirm that the fields Ref Phy Phone and Ref Phy Pager already exist in the database (see <a href="Contextual Search">Contextual Search</a>), otherwise, you must add them. After you add a lookup table, you can add the data to the fields in the table "from scratch," or you can import data from a text file into the fields.

### To add a lookup table

1. On the New Field dialog box, enter the name for the lookup table and select Lookup Table for the field type.

Figure 2-24: New Data Field dialog box, Adding a Lookup Table



- 2. In the Data Folders pane, open the data folder that contains the fields that are to be added to the lookup table.
  - a. The fields that are contained in the data folder are displayed in the Fields pane at the bottom of the Add New Data Field dialog box.
- 3. Drag each field that you are adding to the lookup table from the Fields pane to the Define the Lookup Table Structure pane (upper right pane) of the Add New Data Field dialog box.
- 4. In the Data Folders pane, select the folder where you would like to save your table.

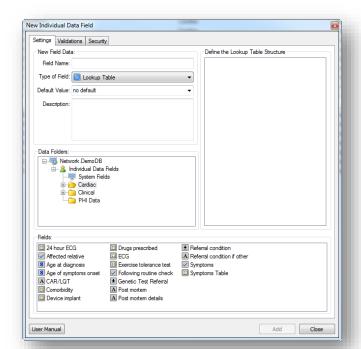


Figure 2-25: New Data Field dialog box, Defining the Lookup Table structure

5. Continue to any other necessary tasks for the field or click Add to add the field the database.

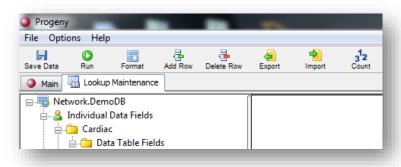
After you have added the lookup table, you can add data to the fields, either "from scratch," or by importing a text file that contains the necessary data. See:

- To add data "from scratch" to a lookup table
- To import data into a lookup table

#### To add data "from scratch" to a lookup table

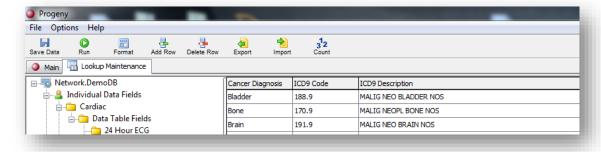
- 1. If you have not already done so, on the navigation bar, click the Fields button to open the Fields window.
- 2. On the main menu, click Fields > Lookup Table Maintenance.
  - a. The Lookup Maintenance tab opens. This tab displays every lookup table (by folder) that currently exists in the selected database.

Figure 2-26: Lookup Maintenance tab



- 3. In the left pane of the Lookup Maintenance tab, select the lookup table to which you are adding data.
- 4. On the Lookup Maintenance toolbar, click the Add Row button Add Row to enter data into the lookup table.

Figure 2-27: Adding data to a lookup table

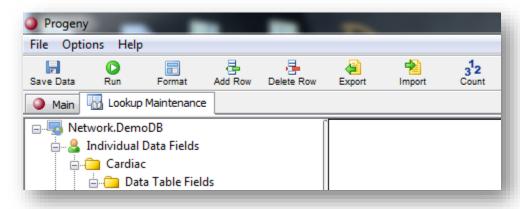


#### To import data into a lookup table

You can import data into a lookup table from a delimited file. The file can be a comma delimited file, a tabdelimited file, or you can specify another delimiter type.

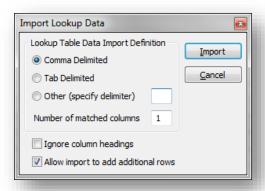
- 1. If you have not already done so, on the navigation bar, click the Fields button to open the Fields window.
- 2. On the main menu, click Fields > Lookup Table Maintenance.
  - a. The Lookup Maintenance tab opens. This tab displays every lookup table (by folder) that currently exists in the selected database.

Figure 2-28: Lookup Maintenance tab



- 3. In the left pane of the Lookup Maintenance tab, select the lookup table into which you are importing the data.
- 4. On the Lookup Maintenance toolbar, click the Import button
  - a. The Import Lookup Data dialog box opens. Comma Delimited is selected by default.

Figure 2-29: Import Lookup Data dialog box



- 5. Select the type of delimited file that you are importing. (If the file is not comma delimited or tab delimited, then you must specify the delimiter that the file uses.)
- 6. Specify the values for the following:
  - a. Number of Matched Columns Enter the number of columns in the text file that you are importing that match to the number of columns in the lookup table. No data in the matched columns is overwritten. Only data from the matched point forward is overwritten. For example, if your lookup table and import text file have three columns, and you indicate "1" for the number of matched columns, then when the data is imported into the lookup table, the data in Column 1 of the lookup table is not overwritten; however, the data in Columns 2 and 3 of the lookup table is overwritten by the data in Columns 2 and 3 of the import file.



The correct value for the number of matched columns is critical to ensuring the integrity of the data in the lookup table. If you are importing a new list into a new table enter a "1" for the number of matched columns.

- b. Ignore column headings—If the file that is being imported contains column headings, select Ignore column headings.
- c. Allow import to add additional rows—To allow the import to add more rows of data to the lookup table during the import process, leave Allow import to add additional rows selected.
- 7. Click Import. The Open File dialog box opens.
- 8. In the Open File dialog box, browse to select and select the file that you are importing, and then click Open. A message opens indicating the Import operation was successful.
- 9. Click OK. The message closes. You return to the Lookup Maintenance tab. The imported information is displayed in the lookup table.
- 10. Position the column widths as you would like them to appear during data entry and select "File | Save Format" from the Main Menu.

# Adding a Computed Field

You cannot enter data into a computed field. Instead, when you add a computed field, you must define an expression for the field that yields a non-editable value that is automatically calculated each time the window that contains the field is opened. For example, a computed field for Current Age is defined as the difference in years between the dates in the Current Date field and the Date of Birth field. The expression can use functions, constants, and values in other fields for the calculation of the value. For example, a computed field can display the Body Mass Index (BMI) for an individual by calculating BMI based on the values in the Height and Weight fields. You can also specify the parameters for a computed field so that the field displays data only if another field is null (no data has been entered in the field). A computed field can be displayed in spreadsheets, pedigrees, and data sheets without displaying the fields on which its calculation is based. For example, the Current Age field can be displayed without displaying the Current Date and/or Date of Birth fields.

### Computed field expressions

An expression is a group of characters or symbols that represent either a quantity/value or an operation. Table 2-2 lists the expressions that are allowed for a computed field.

Table 2-2: Computed field expressions

Expression	Description	Example			
Operators					
<ul> <li>Addition</li> <li>Subtraction</li> <li>Negation</li> <li>Multiplication</li> <li>Division</li> <li>String concatenation</li> </ul> Note: If an expression has a concatenation	<ul> <li>expression + expression</li> <li>expression - expression</li> <li>- (expression)</li> <li>expression * expression</li> <li>expression / expression</li> <li>expression    expression</li> <li>expression + expression</li> </ul>	<ul> <li>SUM = {NUM1} + {NUM2}</li> <li>DIFF = {NUM1} - {NUM2}</li> <li>- (SUM = {NUM1} + {NUM2})</li> <li>PROD = {NUM1} * {NUM2}</li> <li>DIV = {NUM1} / {NUM2}</li> </ul>			
IF THEN ELSE	IF condition THEN expression1 ELSE expression2 ENDIF  Evaluates to expression1 if the condition is true and expression2 if the condition is false. The result is NULL if the condition is NULL or UNKNOWN. You can nest IF conditions.	IF ({NUM1} /500 > 1) THEN 1 ELSE 0 ENDIF			
Note: For detailed information about working with NULL fields with IF/THEN expressions, see NULL values in computed field expressions					
CURRENT DATE CURRENT TIME CURRENT TIMESTAMP NULL CURRENT USER	<ul> <li>All CURRENT constraints put their values from the current client settings</li> <li>A NULL field represents an empty value (not a 0 value)</li> </ul>	<ul> <li>AGE = YEARDIF ({Date of Birth}, CURRENT DATE)</li> <li>DAYS SINCE LAST TREATMENT = DAYS({Treatment Date}, CURRENT_DATE)</li> </ul>			

You can include other computed fields, summary fields, and/or system fields when you are defining the expression for a computed field. For example, in Figure 2-30, the Age database field is a computed field that is used in the calculation of the computed database field named FACTOR1. In Figure 2-31, the Average Age database field is a pedigree Summary field that is used in the calculation of the computed database field named FACTOR2.

Figure 2-30: Including computed fields in the definition of a computed field

AGE = YEARDIF({Date of Birth}, CURRENT DATE)
FACTOR1 = {AGE} \* {Cigarettes Per Day} \* .21321

Figure 2-31: Including Summary fields in the definition of a computed field

An example of including system fields when you are defining the expression for a computed field is the expression {Global ID} + 1000, which is a computed field that would display the value in the system field {Global ID} plus 1000 in the field. (See Appendix A, System Fields)

When you are defining the expression for a computed field, all references to database fields must be enclosed in curly brackets {}. For example, the sum (SUM) of the two database fields NUM1 and NUM2 is defined as {NUM1} + {NUM2}. To make data entry easier, you can drag a database field from the Fields pane at the bottom of the Add New Field dialog box into its correct location in the expression. The database field is automatically enclosed in curly brackets when you add it to the expression.

# NULL values in computed field expressions

A NULL field is a database in which no data has been entered. Any expression that receives a NULL parameter returns NULL, and this is often not an acceptable result. For example, the following expression returns the sum of three numeric fields:

$${Field1} + {Field2} + {Field3}$$

However, if either Field1 or Field2 and/or Field3 is null, the expression returns NULL.

Field Values				
Field1	Field2	Field3	Returned Value	Acceptable Result?
1	2	1	4	Yes
1	NULL	1	NULL	No
NULL	NULL	NULL	NULL	No

You can use the IFNULL function to return a value of one of the fields in the expression, or a zero value. For example, if you use the IFNULL function to define the allowed returned value:

IFNULL({Field1}, 0, {Field1}) + IFNULL({Field2}, 0, {Field2}) + IFNULL({Field3}, 0, {Field3})

then:

	Field Values			
Field1	Field2	Field3	Returned Value	Acceptable Result?
1	2	1	4	Yes
1	NULL	1	2	Yes
NULL	NULL	NULL	0	Yes



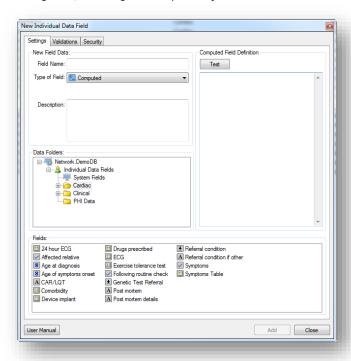
For additional information about the IFNULL function, click <u>here</u> to visit the Sybase website

### To add a computed field

The following example is simply a high-level example of the typical approach used to define a computed field expression. For a complete list of all the functions that are supported by a computed field expression, click <a href="here">here</a> to visit the Sybase website.

1. On the New Field dialog box, enter the name for the computed field and select Computed for the field type.

Figure 2-32: New Data Field dialog box, Adding a Computed field



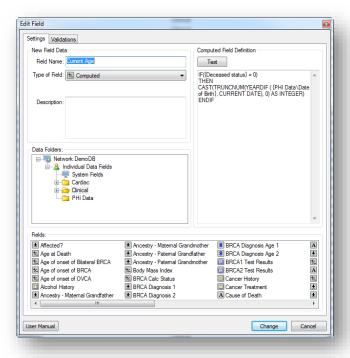
- 2. In the Computed Field Definition pane, enter the expression for the computed field that results in the difference between the Date of Birth and the Current Date (in years) being displayed in the Current Age field. For example:
  - a. Enter the YEARDIF function followed by an open left parenthesis: YEARDIF (
  - b. In the Data Folder pane, open the Demographics folder, and drag and drop the Date of Birth field after the open left parenthesis. The expression now looks like this: YEARDIF ({Demographics\Date of Birth}



Remember, when you drag a database field from the Fields pane at the bottom of the Add New Field dialog box into its correct location in the expression, the database field is automatically enclosed in curly brackets.

After the right curly bracket, enter a comma followed by a space and then the system constant CURRENT DATE. Complete the expression with a closing right parenthesis. The complete expression now looks like this: YEARDIF ({Demographics\Date of Birth}, CURRENT DATE)

Figure 2-33: New Data Field dialog box, Defining the computed field expression



#### 3. Click Test.

- a. If the definition is acceptable, then a message opens stating that the expression is OK.
- b. If the definition is not acceptable, then a message opens indicating that there is an error in the expression.
- Click OK to close the message.
- 4. If the expression is acceptable, then continue to any other necessary tasks for the field or click Add to add the field to the database.
- 5. If the expression has an error, continuing modifying and testing the expression until it is acceptable, then continue to any other necessary tasks for the field, or click Add to add the field to the database.

# Adding a Summary Field

A summary field provides summary information for a given column on a per pedigree basis. Summary functions can be carried out on all fields that are not contained in tables. (The fields can be contained in lookup tables). When you add a summary field, you must specify the type of summary value (Average, Count, Maximum, Minimum, or Sum) that is to be returned in the field.

Summary fields allow you to summarize data from the database field type directly below it – for example:

- · Pedigree-level fields summarize Individual-level data
- · Individual-level fields summarize Sample-level data
- · Sample-level fields summarize Sample-level data (for sub-samples)

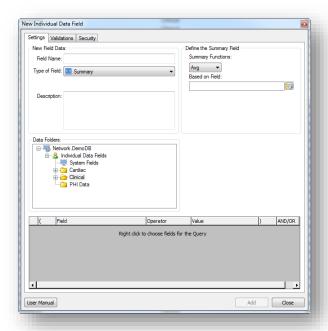


- Summary fields cannot be created at the Marker level
- To summarize the data for a given column in a given table, see <u>Adding a Table</u>
   <u>Summary Field</u>

# To add a summary field

1. On the New Field dialog box, enter the name for the summary field and select Summary for the field type.

Figure 2-34: New Data Field dialog box, Adding a Summary field



- 2. In the Define the Summary Field pane, do the following:
  - a. Select the Summary type. All summaries are calculated on a per pedigree basis. Available values are:
    - i. Avg—Calculate the average value of a field.
    - ii. Count—Return the total count of items in a field.
    - iii. Max—Return the maximum value in a field.
    - iv. Min—Return the minimum value in a field.
    - v. Sum—Return the sum of a numeric field.
  - b. Select the field in the pedigree that you are summarizing.
- 3. Define a query to filter the data:
  - a. Right-click the dark grey area inside the query window and choose Add Row. A field chooser window will open.
  - b. Select the desired field and choose OK.
  - c. Back in the query window, choose the query function and enter the query value.
  - d. Rinse and repeat as necessary.



See <u>Database Query Format</u> for detailed information about formatting a database query.

4. Continue to any other necessary tasks for the field or click Add to add the field the database.

# Adding a Table Summary Field

A table summary field is a computed field that summarizes the data for a given column in a given table. For example, you might add a table called Visit Record with a field in it called Visit Date and you want to see what the most recent visit date is for a specific patient. You can add a table summary field called Most Recent Visit Date that displays the most recent visit date for a patient who has five distinct visit date entries in the Visit Record table.

To summarize the data for a given column in a given pedigree, see Adding a Summary Field

### To add a table summary field

1. On the New Field dialog box, enter the name for the table summary field and select Table Summary for the field type. Choose the table field you want to summarize and click OK.

Figure 2-40: Select Table dialog box

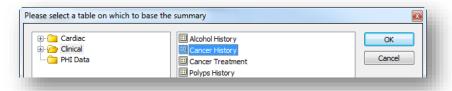
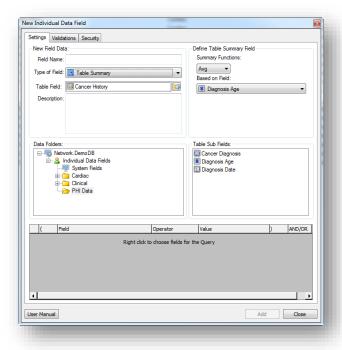


Figure 2-41: New Data Field dialog box, Adding a Table Summary field



- 2. In the Define Table Summary field pane, select the Summary Function. Available values are:
  - a. Avg Calculate the average value of a field.
  - b. Count Return the total count of items in a field.
  - c. Max Return the maximum value in a field.
  - d. Min Return the minimum value in a field.
  - e. Sum Return the sum of a numeric field.
- 3. Select the field in the table that you are summarizing from the Based-on Field drop-down.
  - a. If you choose the Count function, this dropdown will disappear. Count functions are configured entirely in the Query window.
- 4. Optionally you can define a query to further filter the data:
  - a. Drag a field (you can select multiple subfields by using CTRL + click) from the Table SubFields pane to the Query pane then choose a Query Operator type and enter the Query Value.



See <u>Database Query Format</u> for detailed information about formatting a database query.

In Figure 2-43 below, the table summary field counts the number of rows in the Cancer History table and uses the Query window to restrict this count to only table rows where the Cancer Diagnosis subfield has a value of "Breast."

New Individual Data Field

Settings Validations Security

New Field Data:

Field Name: Breast Cancer Count!

Type of Field: Table Summay

Table Field: Cancer History

Description:

Data Folders:

Table Sub Fields:

System Fields

System Fields

Field Operator Value

Field Operator Value

Cancer Diagnosis

Field Operator Value

AND/OR

Figure 2-43: Adding a Table Summary field, Defining the field

User Manual

5. Continue to any other necessary tasks for the field or click Add to add the field the database.

Add Close

# Copying Fields between Databases

When you are creating a new database or modifying an existing database, you have the option of adding new fields "from scratch" to the database; however, if an existing database already contains the needed fields, you can simply copy these fields. When you copy fields between databases, if any fields have the same name and are of the same or different type, then a prompt opens asking you if you want to overwrite the original field and all the data in the field. You can select to overwrite the original field and all its data, or you can leave the original field and its data as-is.

### To copy fields between databases

- 1. Log in to the database from which you are copying the fields.
- 2. On the Progeny main window navigation bar, click the Fields button . The Fields window opens.
- 3. If you are copying all the data fields for all the record types, go to Step 4; otherwise, do one of the following:
  - a. To copy only selected fields, in the left pane of the Fields window, open the folder that contains the fields that are to be copied, and then in the right pane of the main window, select the fields (CTRL-click to select multiple fields).
  - b. To copy all the fields for a record type, in the left pane of the Fields window, select the record type. For example, to copy all the Individual data fields, then select Individual Data Fields.
- 4. On the main menu, click Fields > Save Fields as Text, and then select one of the following:
  - a. Save Selected Fields.
  - b. Save All < > Data Fields, where < > can be Individual, Pedigree, Sample Data, or Marker Data, for example, Save All Individual Data Fields.
  - c. Save All Data Fields.



Select Save All Data Fields only if you are copying all the data fields for all the record types.

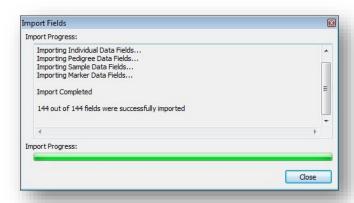
- 5. The selected fields are saved to a file and the Open dialog box opens.
- 6. In the Open dialog box, browse to the location where the copied fields are to be saved, enter a name for the file that contains the copied fields, and then click Open.



#### The export file will be saved as a text file – users cannot change this file type.

- 7. Log out of the database from which you are copying the fields, and then log in to the database into which you are copying the fields.
- 8. On the navigation bar, click the Fields button . The Fields window opens.
- 9. On the main menu, click Fields > Load Fields from Text. The Open dialog box opens.
- 10. Browse to and select the text file that you saved, and then click Open.
  - a. If none of the fields in the text file has the same name as any of the fields in the database into which you are importing the fields, then the Import Fields dialog box opens. The dialog box indicates the number and type of fields that were successfully imported into the database; otherwise, for each matching field, a prompt opens that asks you if you want to overwrite the original field and all its data. Click Yes to overwrite the original field and all its data or click No to leave the original field and all its data as-is. After you have responded to the prompt for each matching field, the Import Fields dialog box opens.

Figure 2-44: Import Fields dialog box



11. When the import is complete, click Close to close the Import Fields dialog box.

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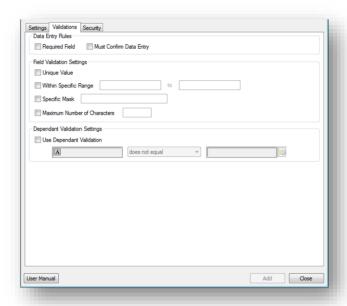
# Setting Validations for a Database Field

When you are adding or editing a database field, you have the option of setting validations for the field. Validations guarantees to the Progeny that every data value that is entered the field is correct and accurate. Three types of field-level validations are available—data entry rules, field validation settings, and dependent validations. If field validation settings and/or dependent validations are violated when you attempt to save data, then validation errors are generated.

### To set validations for a database field

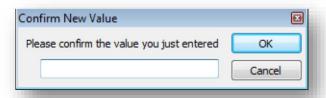
- 1. If the field is already open in the Add New Field dialog box, then go to Step 3; otherwise, on the navigation bar, click the Fields button it to open the Fields window.
- 2. In the left pane of the Fields window, open the folder that contains the field for which you are setting the validations, and then in the right pane of the window, double-click the field to open the field in the Edit Field dialog box.
- 3. Open the Validations tab on the Add New Field/Edit Field dialog box.

Figure 2-45: Add Data Field dialog box, Validations tab



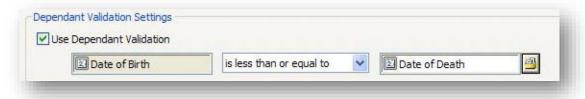
- 4. Set the validations for the field.
  - a. Data Entry Rules—These rules are applied only at data entry time. (Data entry in a datasheet, spreadsheet, and the Update Workflow window.)
  - b. Required Field—You must enter a value in the field before you can the data.
  - c. Must Confirm Data Entry—You are prompted to re-enter the data in the field before you can save the data. If a field has the Must Confirm Data Entry validation assigned to it, then a Confirm New Value dialog box opens for repeating the entry.

Figure 2-46: Confirm New Value dialog box



- d. Field Validation Settings—These settings are applied at the database level and are applied always (data entry, imports, and so on).
- e. Unique value—You must enter a unique value in the field wherever it is used (pedigree, individual, or sample).
- f. Within Specific Range—Indicates the allowed range of values (numbers, dates, and so on) for the field.
- g. Specific Mask—Defines how you must enter data into the field maintain consistency and to make your database easier to manage. For example, with the underscore character (\_) representing a single character, an input mask of (\_\_\_)\_\_\_- would require a phone number like this: (574)968-0822.
- h. Maximum Number of Characters—Indicates the maximum number of characters that you can enter the field.
- i. Dependent Validation Settings—These settings are applied at the database level and are applied always (data entry, imports, and so on).
- j. Select Use-Dependent Validation, and then specify the relation that the field must have with another field of the same type. For example, the following figure shows how the value in the Date of Birth field must be less than or equal to the value in the Date of Death field.

Figure 2-47: Example of a dependent validation setting



5. Continue to any other necessary tasks for the field or click Add to add the field the database.

#### Data validation errors

If field validation settings and/or dependent validations are violated when you attempt to save data, then the fields with the validation errors are filled with red on the affected data sheet and spreadsheet and a Validation Errors dialog box opens. This dialog box lists the of validation errors on a field by field basis. You can double-click on a field that is listed in error to go the field in the open datasheet or spreadsheet.



Data entry rules are not included in the generation of validation errors. Instead, you are prompted with the rules before you save the data.

Figure 2-48: Data validation errors noted on a datasheet

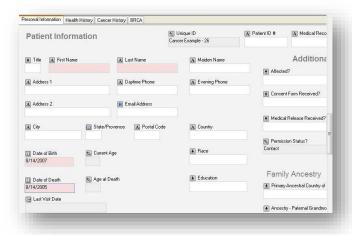


Figure 2-49: Data validation errors noted on a spreadsheet

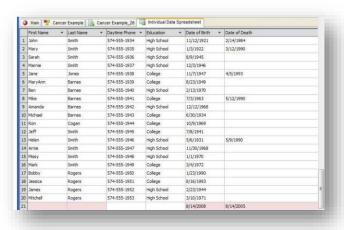
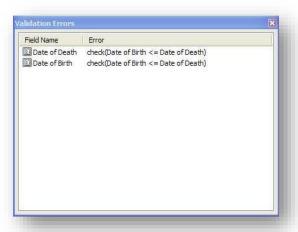


Figure 2-50: Validation Error dialog box



You can double-click on a field that is listed in this dialog box to go the field in the open datasheet or spreadsheet.

# Adding, Editing and Deleting Database Folders

Folders provide a means of organizing your database fields. You can add as many root folders and sub-folders as needed to support your business needs. After you add a folder, you can set the security for the folder as well as for fields that are in the folder. You can also edit the security for a database folder. The default security settings for a folder are set to Read and Write for All Users. As the Progeny administrator, if you have yet to add any other user classes, then only two default user classes—Clinical and Laboratory—are displayed for a folder; otherwise, all the user classes that you have added are displayed. You can add more classes if needed. You can delete a folder only if the folder is empty (contains no database fields).

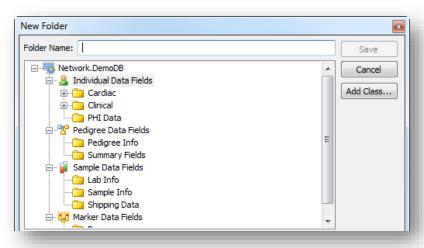


For information about adding a user class and setting folder security for a user class, see Setting Database Folder Security

#### To add a database folder

- 1. On the navigation bar, click the Fields button to open the Fields window.
- 2. On the Fields window toolbar, click the New Folder button New Folder. The New Folder dialog box opens.

Figure 2-51: New Folder dialog box



3. In the Folder Name field, enter the name for the new folder.

4. Select the location for the new folder. For example, to create a folder at the root level for Individual data, select Individual Data Fields. To create a folder that is a subfolder of Individual Demographic data, under Individual Data Fields, select the Demographics folder.



You can create as many root folders and subfolders as needed to support your business needs.

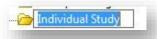
- 5. Click Save. A message opens indicating that the folder was successfully added.
- 6. Click OK. The message closes. You return to the Fields window. The newly added folder is displayed in the window.
  - a. When you create database fields, you typically set the security for the fields on a field by field basis; however, if you want to apply the same security settings for every single field that is contained in the same database folder in a single step, then you can apply the settings at the folder level. See Setting Database Folder Security

### To edit a folder

You can rename a folder, and you can change the security settings for a folder.

1. To rename a folder, triple-click on the folder in the Fields window to select it and then modify the name as needed.

Figure 2-52: Example of a folder name selected for modifying



2. To modify the security for a folder, see <u>Setting Database Folder Security</u>

## To delete a database folder

You can delete a selected database folder only if the folder is empty (contains no database fields). If you want to delete a database folder that contains fields, you must first delete all the fields in the folder. (See <u>To delete a database field</u>)

- 1. On the navigation bar, click the Fields button to open the Fields window.
- 2. In the left pane of the Fields window, right-click on the folder that you are deleting, and on the context menu that opens, select Delete Folder.
  - a. A message opens asking you to confirm the deletion of the folder.
- 3. Click OK to close the message and delete the folder

## Chapter 3 – Datasheet Basics

A datasheet is a paper form or document that details the properties of a product or component. The form contains fields for collecting specific data about the product or component. By analogy, you use datasheets in Progeny to enter and store data for individuals, pedigrees, markers, and samples. Each datasheet contains database fields that store specific data for an individual, a pedigree, a marker set, and or a sample.

This chapter covers the following topics:

- Datasheet Types
- Creating and Formatting Datasheets
- Working with Tables in a Datasheet
- Saving and Loading Datasheet Formats
- Printing a Datasheet



This chapter addresses only the basics for all datasheet types. Detailed information about adding the necessary components for creating a specific type of datasheet (for example, creating folders and adding individuals to the folder so that you can create an individual datasheet) are discussed in detail in the appropriate chapter (Progeny Clinical, Progeny Lab, or Progeny LIMS) in this manual.

# **Datasheet Types**

There are five distinct types of datasheet that can be created in Progeny:

- 1. Individual Datasheets The individual datasheet is used to enter and store information about individuals in your Progeny database. An Individual datasheet can contain individual database fields, pedigree database fields, and marker database fields. Link to detail
- 2. Pedigree Datasheets The pedigree datasheet is used to enter and store information about pedigrees in your Progeny database. A Pedigree datasheet can contain only pedigree database fields. Link to Detail
- 3. Sample Datasheets The sample datasheet is used to enter and store information about samples in your Progeny database. Additionally, the sample datasheet is the only one of the five datasheet types that can contain all four types of Progeny database fields—individual database fields, pedigree database fields, sample database fields, and marker database fields. Link to Detail
- 4. Marker Datasheets The marker datasheet is used to enter and store information about markers in your Progeny database. A Marker datasheet can contain only marker database fields. Link to Detail
- 5. Table Datasheets The table datasheet is a special type of datasheet as the scope of fields that can be added to it are limited to the subfields of a table. Each table datasheet can contain table subfields from only a single table. A table datasheet is, in turn, contained in one of the other four types of datasheets individual, pedigree, sample, or marker. Link to Detail

## **Creating and Formatting Datasheets**

After adding the necessary field types to your Progeny database, and you have added the necessary components for creating a specific type of datasheet (for example, you have created Individual folders and you have added individuals to a folder in preparation for creating an individual datasheet), you can create the datasheet. When you are creating a datasheet, you have a variety of options for formatting the datasheet, including the following:

- Adding tabs to the datasheet.
- Positioning and aligning the fields on the datasheet.
- Specifying default settings for the fields on the datasheet.
- Adding static text to the datasheet.
- Creating table datasheets for table fields.



The following procedure assumes that you have already added the required components for creating a specific type of datasheet. (For example, you have created folders and added individuals to the folder so that you can create an individual datasheet.) Detailed procedures for creating the necessary components for adding a specific type of datasheet are discussed in detail in the appropriate chapter (Progeny Clinical, Progeny Lab, or Progeny LIMS) in this manual.

## To create and format a datasheet

1. At the top of the blank datasheet, click Form Design.

Figure 3-1: Datasheet toolbar

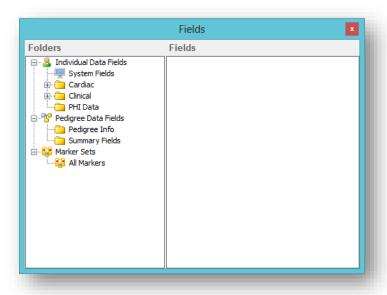


2. On the datasheet toolbar, click the Fields button to open the Fields dialog box.



3. This dialog box contains all the fields (grouped by folder) that you can add to the datasheet. It also lists the system fields that you can add to the datasheet. For example, if you are creating an individual datasheet, then the dialog box lists all the individual data fields (grouped by folders), all the pedigree data fields (grouped by folders), and all the marker sets (grouped by folder) that you can add to the datasheet. It also lists all the individual system fields that you can add to the datasheet. See Figure 3-2 below.

Figure 3-2: Fields dialog box for an Individual datasheet



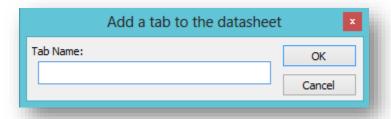


System fields are read-only fields that Progeny automatically stores in its database for a record. Each record type (individual, pedigree, sample, and marker) has its own unique system fields. You can include system fields on a datasheet if there is information that you want to view always, but not edit, for an individual, a pedigree, a sample, or a marker. For a complete list of system fields and the read-only value that is displayed in each field, see <u>Appendix A – System Fields</u>

4. If the datasheet is to be a single sheet, (that is, no tabs), then continue to Step 5; otherwise, for each tab that you are adding to the datasheet, on the datasheet toolbar, click the Add Tab button to open the Add Tab dialog box, enter a name for the new tab, and then click OK.

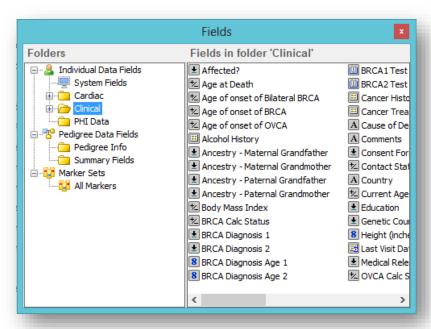


Figure 3-3: Add Tab dialog box



5. In the left pane of the Fields dialog box, open the folder that contains the field or fields that you are adding to the datasheet. The list of fields contained in the folder is displayed in the right pane of the Fields dialog box.

Figure 3-4: Adding Fields (Example shown here: using Clinical Folder)

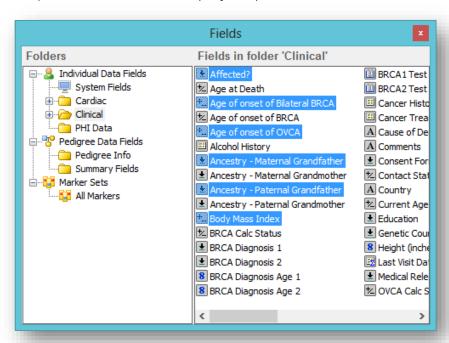




If you need to review the different field types that you can add to a datasheet, see Adding a Table

6. If applicable, make sure that the correct tab is open and then from the right pane of the Fields dialog box, drag the needed field or fields (CTRL-click to select multiple fields) to the datasheet.

Figure 3-5: Adding Fields (CTRL-click to select multiple fields)





- You can add the same field to multiple tabs on the datasheet. The field can be shown in different locations on each tab if needed. When you enter data in a field on one tab, the same data is automatically entered in the field on the other tabs.
- If you added a field in error, or want to remove a field from the datasheet, select the field, and then click the Delete key on your keyboard.
- 7. Format the fields on your datasheet as necessary. See:
  - a. To align multiple fields
  - b. To add static text to a datasheet
  - c. To set the properties for all the fields on the datasheet
  - d. To set the field properties on a field by field basis
  - e. Working with Tables in a Datasheet

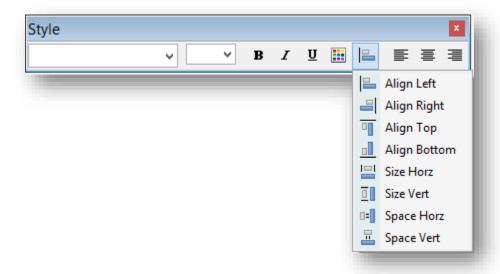
## To align multiple fields

- 1. CTRL-click to select the fields that you are aligning.
- 2. On the datasheet toolbar, click the Style button to open the Style toolbar.



3. On the Style toolbar, click the Alignment button to open a menu with a variety of options for modifying the field alignment.

Figure 3-6: Style toolbar



## Alignment Button Options

Option	Description
Align Left	From top to bottom, vertically aligns the left edges of the selected fields.
Align Right	From top to bottom, vertically aligns the right edges of the selected fields.
Align Top	From left to right, horizontally aligns the top edges of the selected fields.
Align Bottom	From left to right, horizontally aligns the bottom edges of the selected fields.
Size Horz	Sets the width of all the selected fields to the same width as the first selected field.
Size Vert	Sets the height of all the selected fields to the same height as the first selected field.
Space Horz	Sets the horizontal spacing between all the selected fields to the same as the spacing between the first two selected fields.
Space Vert	Sets the vertical spacing between all the selected fields to same as the spacing between the first two selected fields.

## To add static text to a datasheet

Static text is free-standing text that is not tied to any field on a datasheet. You can add static text anywhere on a datasheet.

1. On the datasheet toolbar, click the Add Text button.



2. A blank text box with the phrase "Static Text" is displayed on the worksheet.

Figure 3-7: Blank text box



- 3. Click on the phrase "Static Text" to select it, and then replace the phrase with the appropriate text.
- 4. Drag the top, side, or corner handles to resize the text box as needed.
- 5. Click and hold on the center of the text box and drag the text box to the correct location on the datasheet.
- 6. To change the format for the static text (font size, font color, and so on), see one of the following:
  - To set the properties for all the fields on the datasheet
  - To set the field properties on a field by field basis

## To set the properties for all the fields on the datasheet

You can set the same default properties for all the fields on the datasheet and all static text in a single step.

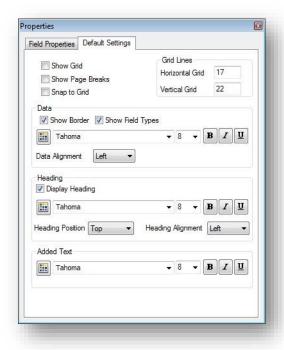
1. On the datasheet toolbar, click the Properties button to open the Properties dialog box.

2.



3. If needed, open the Default Settings tab.

Figure 3-8: Properties dialog box, Default Settings tab



4. Set the default values as needed.

Option	Description
Show Grid	Displays a background grid (a set of intersecting lines used to align objects) on the datasheet.
Show Page Breaks	Displays the edges of the print pages in Print Preview mode.
Snap to Grid	When moving a field, aligns (or "snaps") the field to the nearest intersection of lines in the grid, even if the grid is not visible.
Data	Show Border—Shows the border around all fields.  Show Field Type—Displays the icon for each field at the top left corner of the field.  Data Alignment—The horizontal alignment of the data that is displayed in the fields. Values are Left, Center, and Right.
Heading	Display Heading—Displays the field name as the field heading for each field.  Heading Position—The location of the field heading relative to the field. Values are Top, Left, Bottom, and Right.  Heading Alignment—The alignment of the field heading relative to the field. Values are Left (aligned with the left edge of the field), Center (centered relative to the field), and Right (aligned with the right edge of the field.



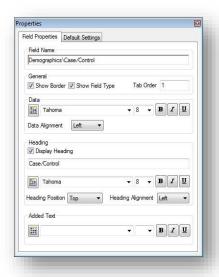
Use the formatting options to format the font color, font type, font size, and font properties (Bold, Italics, or Underline) for the data that is entered in a field, the field headings, and any static text on the datasheet.

## To set the field properties on a field by field basis

You can set the properties for all fields on the datasheet, including static text fields, on a field by field basis.

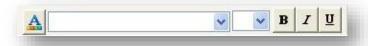
- 1. Select the field for which you are setting the properties.
- 2. On the datasheet toolbar, click the Properties button Properties to open the Properties dialog box.
- 3. If needed, open the Field Properties tab. The database name of the selected field (including the name of its folder) is displayed in the Field Name field and you cannot change this value.

Figure 3-9: Properties dialog box, tab



4. Set the default values as needed.

Option	Description
General	Show Border – Shows the border around the field.  Show Field Type – Displays the icon for the field at the top left corner of the field.  Tab Order – The order in which a user tabs through the fields on a datasheet. The default tab order is the order in which you added the fields to the datasheet.
Data	Data Alignment – The horizontal alignment of the data that is displayed in the field. Values are Left, Center, and Right.
Heading	Display Heading – Displays a user-defined value as the heading for the field.  Note: Any value that you enter here does not overwrite the database name for the field. It is simply a display value for the field on the datasheet.  Heading Position – The location of the field heading relative to the field. Values are Top, Left, Bottom, and Right.  Heading Alignment – The alignment of the field heading relative to the field. Values are Left (aligned with the left edge of the field), Center (centered relative to the field), and Right (aligned with the right edge of the field.



Use the formatting options to format the font color, font type, font size, and font properties (Bold, Italics, or Underline) for the data that is entered in the field, the field heading, or any static text on the datasheet.

# Working with Tables in a Datasheet

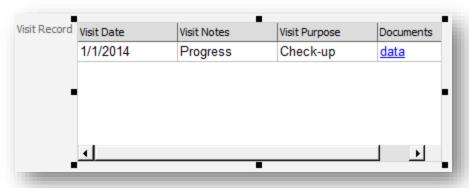
Data entry for most of the fields in a datasheet is very straightforward and is described for each field type in <a href="Database Field Types">Database Field Types</a>. The tab order for the fields determines the order in which you move through the fields on the datasheet and enter data in them. A variety of options, however, are available for working with table fields in datasheets, including <a href="resizing a table">resizing a table</a>, <a href="Saving and loading table formats">Saving and loading table formats</a>, and <a href="entering data either directly into the table">entering table datasheets</a> and entering the data into a table through its table datasheet.

#### To resize a table

When adding a table field to a datasheet, the table is added with a default size (width and height). Resize the window to suit your specific needs.

- 1. At the top of the datasheet, click Form Design.
- 2. Click in the table field to select it.
- 3. Resizing handles appear on the table.

Figure 3-10: Selected table in a datasheet



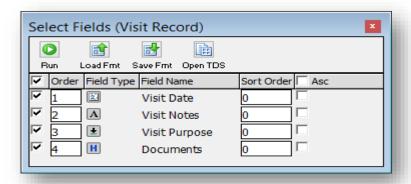
4. Drag the top, side, or corner handles to resize the table as needed.

#### To format a table in a datasheet

When adding a table to a datasheet, the order in which the fields are displayed in the Define the Table Structure pane in the Add New Field dialog box (see <u>Adding a Table</u>) is the order in which the rows are displayed in the table in the datasheet. It is possible to change this order and the new order can be saved as a table format. You can save multiple formats for the same table and load different formats for the same table in different datasheets (but you can only have one format loaded per table per datasheet).

- 1. In Form Design mode, right-click on the table in the datasheet, and on the context menu that opens, click Show Fields.
  - a. The Select Fields window opens. This window lists all the fields in the table and their current order in the table.

Figure 3-11: Select Fields table



- 2. In the Order column, enter the new order for the table fields.
- 3. Optionally, to sort the table display, enter the sort order for the table fields.
  - a. For example, to sort the data in the Visit Table shown in Figure 3-9 above by the most recent visit date, in the Sort Order column for Visit Date, enter a "1," and then select Asc (which indicates Ascending order). To sort by the oldest visit data, do not select Asc.

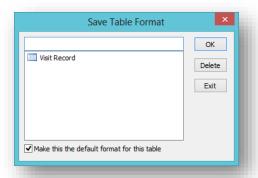


4. Click the Run button Run to change the order of the table fields in the datasheet.



- 5. Optionally, to save this new order of rows as a table format, click the Save Format button Save Fmt
  - a. The Save Table Format dialog box opens.

Figure 3-12: Save Table Format dialog box



6. Enter a name for the table format, and if you want to make this format the default format for the table, leave the "Make this the default format for the table" checkbox selected.



An unlimited number of formats can be created, but only one format can be set as the default format per table within a datasheet.

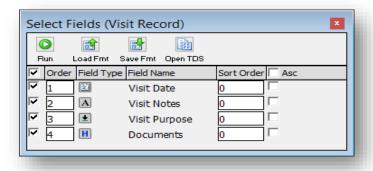
7. Click OK to commit your choice and exit the Save Table Format window.

### To load a different format for a table in a datasheet

When adding a table field with multiple formats to a datasheet, the table is added in its default format. You can load a different format for the table.

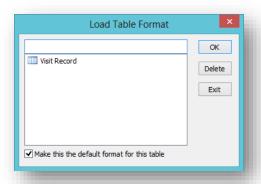
1. In Form Design mode, right-click on the table in the datasheet, and on the context menu that opens, click Show Fields. The Select Fields window opens. This window lists all the fields in the table and their current order in the table.

Figure 3-13: Select Fields table



2. Click the Load Format button Load Fmt. The Load Table Format dialog box opens. This dialog box lists all the available formats for the table.

Figure 3-14: Load Table Format dialog box



a. Select the format that you want to apply to the table, and then click OK. The Load Table Format dialog box closes, and you return to the Select Format window.

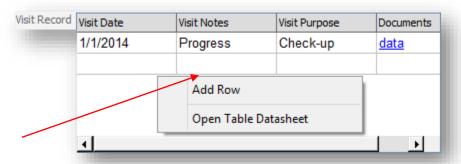


3. Click the Run button Run to apply the selected format to the table.

## To enter data directly into a table in a datasheet

- 1. At the top of the datasheet, click Data Entry.
- 2. Right-click in the table, and on the context menu that opens, click Add Row.
  - a. A new blank row opens for adding data.

Figure 3-15: Table with a blank row for adding data



3. Click in each table cell of the blank row to add the necessary data.



If a table has many columns, you can create a table datasheet for the table to make data entry easier.

### To create a table datasheet

If you have a table in a datasheet that has many fields in which to enter data, then date entry can become quite cumbersome. To make data entry easier for a large table, you can set up a table datasheet for the table. The table datasheet displays all the fields for a table row in a single view. You do not have to scroll through a table row to add data to each field in the row. You can create a table datasheet from both the Form Design mode and the Data Entry mode.

#### To create a table datasheet while in Form Design mode

- 1. Right-click on the table, and on the context menu that opens, click Show Fields.
  - a. The Select Fields window opens. This window lists all the fields in the table and their current order in the table.

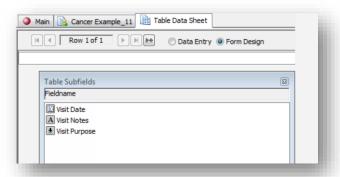
Figure 3-16: Select Fields table





- 2. Click the Open TDS button Open TDS.
  - a. The Table Subfields dialog box opens. This dialog box lists all the fields in the table.

Figure 3-17: Table Subfields dialog box

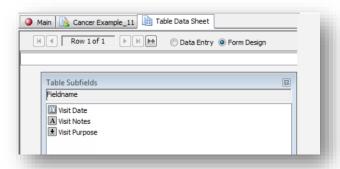


- 3. Drag the needed field or fields (CTRL-click to select multiple fields) to the datasheet.
- 4. Optionally, format the fields. See:
  - a. To align multiple fields
  - b. To add static text to a datasheet
  - c. To set the properties for all the fields on the datasheet
  - d. To set the field properties on a field by field basis
- 5. Continue with any other activities as needed for the datasheet, and after the datasheet is saved, you can enter data into the table datasheet. See <u>To enter data into a table datasheet</u>

#### To create a table datasheet in Data Entry mode

- 1. Right-click on the table, and on the context menu that opens, click Open Table Datasheet.
  - a. The Table Subfields dialog box opens. This dialog box lists all the fields in the table. The mode changes to Form Design.

Figure 3-18: Table Subfields dialog box



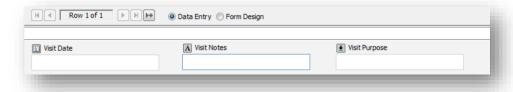
- 2. Drag the needed field or fields (CTRL-click to select multiple fields) to the datasheet.
- 3. Optionally, format the fields. See:
  - a. To align multiple fields
  - b. To add static text to a datasheet
  - c. To set the properties for all the fields on the datasheet
  - d. To set the field properties on a field by field basis

You now have added the first blank row for the table datasheet, and you can continue immediately with entering data in the table datasheet. Go to Step 2 to enter data into a table datasheet below.

## To enter data into a table datasheet

- 1. In Data Entry mode, right-click on the table, and on the context menu that opens, click Open Data Table Sheet.
  - a. A blank row in the table datasheet opens.

Figure 3-19: Example of a blank row in a table datasheet



- 2. Enter the data in the table row.
- 3. Click the Save Data button Save Data.
- 4. To add a new data row, click the New Row button.
- 5. Repeat Steps 2-4 until all the necessary data is added to the table



To move among the different rows in the table datasheet, use the navigation buttons at the top of the datasheet.

Button	Description
H	Move to the first row in table datasheet.
4	Move to the previous row in table datasheet.
<b>F</b>	Move to next row in table datasheet.
M	Move to the last row in table datasheet.

# Saving and Loading Datasheet Formats

After you have created a datasheet with a specific format, you can save the format. You can then retrieve this saved format and load it into another datasheet of the same type.

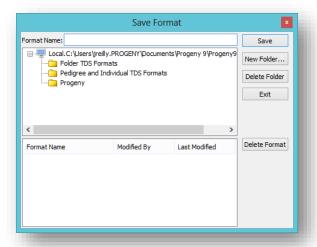
### To save a datasheet format

1. Create and format the datasheet as needed. See Creating and Formatting Datasheets



- 2. On the datasheet toolbar, click the Save Format button Save Fmt.
  - a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the datasheet format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 3-20: Save Format dialog box



- 3. Enter a name for the datasheet format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
  - c. Enter the name of the new folder in which to save to format.
  - d. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
- 4. Click Save on the Save Format dialog box to commit the format and close the Save Format window.

## To load a datasheet format

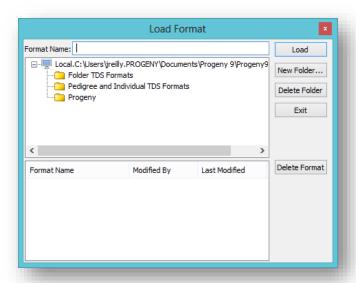
You can a retrieve a saved datasheet format and load it into another datasheet of the same type.

1. Open the datasheet into which you are loading a saved format.



- 2. On the datasheet toolbar, click the Load Format button Load Fmt .
  - a. The Load Format dialog box opens.

Figure 3-21: Load Format dialog box



- 3. Open the folder that contains the datasheet format that you are loading, select the format, and then click Load.
  - a. The datasheet is refreshed with the loaded format. You can now edit the datasheet, enter data, and so on.

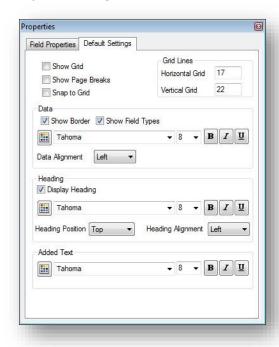
# Printing a Datasheet

You can print a datasheet in either portrait or landscape mode. You can always view the page breaks for multiple page datasheets before you print the datasheet.

## To print a datasheet

- 1. Optionally, before you print the datasheet, to view the page breaks on each page, do the following:
  - At the top of the datasheet, click Form Design.
  - On the datasheet toolbar, click the Properties button Properties to open the Properties dialog box.
  - If needed, open the Default Settings tab.
  - Select Show Page Breaks.

Figure 3-22: Properties dialog box, Default Settings tab



- 2. Optionally, right-click on the datasheet and on the context menu that opens, click Print Setup to open the Print Setup dialog box and confirm or modify the printing options as needed, including select the printing orientation (Portrait or Landscape).
- 3. On the datasheet toolbar, click the Print button Print

# Chapter 4 Spreadsheet Basics

A spreadsheet is a table of values that are arranged in rows and columns. In Progeny, you use spreadsheets to organize multiple individuals, pedigrees, samples, or markers by rows and the database fields for entering the corresponding data by columns.

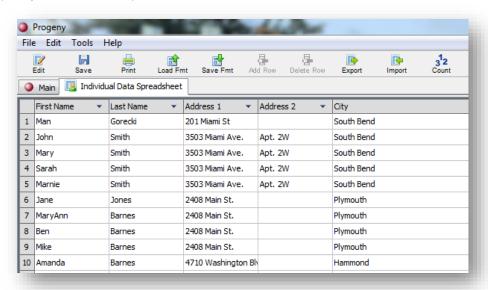
This chapter covers the following topics:

- Spreadsheet Types
- Generating a Spreadsheet
- Entering Data into a Spreadsheet
- Exporting Spreadsheet Data
- Saving and Loading a Spreadsheet Format
- Running Spreadsheet Queries
- Sorting and Filtering Spreadsheet Data
- Printing Spreadsheet Data
- Counting Spreadsheet Data

# **Spreadsheet Types**

A spreadsheet is a table of values that are arranged in rows and columns. In Progeny, you use spreadsheets to organize your individuals, pedigrees, samples, or markers by rows and the database fields for entering the corresponding data by columns.

Figure 4-1: Example of an individual spreadsheet



Unlike a datasheet, which displays the data for a single individual, pedigree, sample, or marker, a spreadsheet displays the data for all individuals, all pedigrees, all samples or all markers in a Progeny database.

You can create four different types of spreadsheets in Progeny:

- <u>Individual spreadsheets</u> Individual spreadsheets are used to enter and store information about individuals in your Progeny database. An individual spreadsheet can contain individual database fields, pedigree database fields, and marker database fields.
- <u>Pedigree spreadsheets</u> Pedigree datasheets are used to enter and store information about pedigrees in your Progeny database. A Pedigree datasheet can contain only pedigree database fields, including summary fields.
- <u>Sample spreadsheets</u> Sample spreadsheets are used to enter and store information about samples in your Progeny database. A Sample datasheet can contain all four types of Progeny database fields—individual database fields, pedigree database fields, sample database fields, and marker database fields.
- <u>Marker spreadsheets</u> Marker spreadsheets are used to store information about markers in your Progeny database. A Marker datasheet can contain only marker database fields.

# Generating a Spreadsheet

Unlike a datasheet, which displays the data for a single individual, pedigree, sample, or marker, a spreadsheet that you generate from the Progeny main window displays the data for all individuals, all pedigrees, all samples or all markers in a Progeny database. A spreadsheet that is generated for all individuals, all pedigrees, and so on in a Progeny database is sometimes referred to as a global spreadsheet.



It is possible to generate a spreadsheet just for the individuals who are contained in the same pedigree. See <u>Generating a Spreadsheet for the Individuals in a</u>

Pedigree

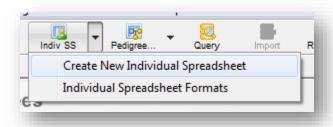
## To generate a spreadsheet



The following procedure details the generation of an Individual spreadsheet, but, by analogy, this procedure can be applied to generate spreadsheets for all pedigrees, all samples, and all markers in a Progeny database.

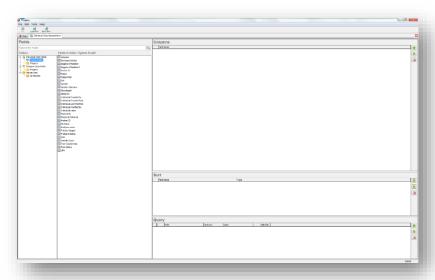
1. From the Progeny Main Screen, click the down arrow next to the Indiv SS button. You can choose either Create New Individual Spreadsheet or Individual Spreadsheet Formats.

Figure 4-2: Main menu Individual Spreadsheet launch options



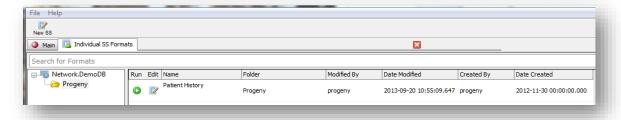
a. If you choose, Create New Individual Spreadsheet, a new tab opens with Fields, Columns, Sort and Query windows.

Figure 4-3: Fields dialog box and Spreadsheet Format window

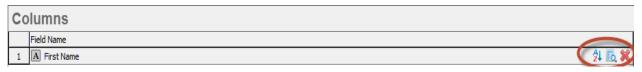


b. If you choose Individual Spreadsheet Formats, a new tab opens allowing you to search for, edit and run saved spreadsheet formats. For additional information on this menu, please see <u>To load a spreadsheet format</u>.

Figure 4-4: Individual Spreadsheet Formats menu



- 2. In the Create New Individual Spreadsheet menu, drag fields into the Columns, Sort, or Query Windows.
  - a. The search bar above the field chooser on the left provides a full-text search across your fields. You can enter a whole or partial field name and the search function will display all possible matches.
  - b. If you add a field to the Columns pane and hover over its row, you will see these three icons appear on the right side:



- i. Adds the row to the Sort pane.
- ii. Adds the row to the Query pane.
- ii. Deletes the row from the Columns pane.
- c. To the right of the Columns, Sort and Query panes are three buttons stacked vertically:



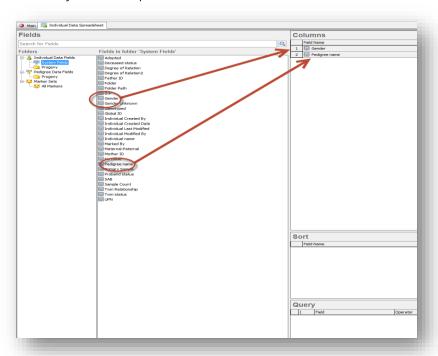
- i. Moves the selected field/row up one position in the order
- ii. Moves the selected field/row down one position in the order
- iii. A so-called "Nuclear Option," this button will delete all fields/rows within the selected pane.



System fields are read-only fields that Progeny automatically stores in its database for a record. Each record type (individual, pedigree, sample, and marker) has its own unique system fields. System fields can be included in a spreadsheet if there is information that needs to be visible always, but not edits, for an individual, a pedigree, a sample, or a marker. For a complete list of system fields and the read-only value that is displayed in each field, see Appendix A, System Fields

For all fields other than table fields, the order in which the fields are dragged is the order in which they appear in columns (from left to right) in the spreadsheet. If you wish to include table fields in your spreadsheet, please see the <a href="Tables in spreadsheets">Tables in spreadsheets</a> section below. If you wish to include a lookup table field in your spreadsheet, please see the <a href="Lookup tables in spreadsheets">Lookup tables in spreadsheets</a> section below.

Figure 4-5: Dragging non-table fields to a spreadsheet



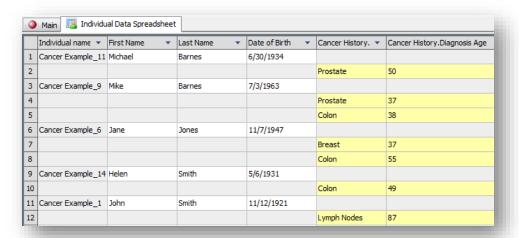
- 3. Optionally, to sort the data in the spreadsheet, drag fields from the left side into the Sort pane or click the Sort button on one or more of the rows in your Columns pane.
  - a. Use the and buttons to change the sort order.
  - b. For each row, in the sort order, you can choose between Ascending (A-Z) or Descending (Z-A) order.

Figure 4-6: Configuring the Sort pane when building an Individual Spreadsheet



4. Click the Run button to generate the spreadsheet. If any data is entered the spreadsheet, click the Save button save to commit the changes. Lastly, if you have run the spreadsheet and want to make changes to the columns, sort order or query click the Edit button to return to the Create New Individual Spreadsheet menu.

Figure 4-7: Individual Spreadsheet Example





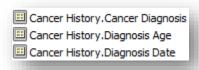
If you need to adjust the width of a spreadsheet column, place the mouse cursor on the black line inside the column heading separating the column you want to adjust and the column immediately to its right. The cursor will change to a double-headed arrow indicating that you can drag the column to the left or right.

5. If you want to view the pedigree of an individual within the spreadsheet or see an individual's datasheet, right click on the first column displayed for that person's row and choose either Open Pedigree or Open Individual Datasheet.

#### Tables in spreadsheets

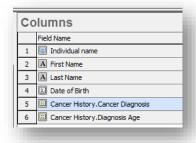
Progeny 9 changes how tables are handled within the spreadsheet modules. When you build a spreadsheet, tables are no longer displayed as an individual entity but are broken apart with each of their table subfields being displayed as a distinct entity.

Figure 4-8: A table displayed as a group of subfields within the Create New Individual Spreadsheet menu.



In previous versions of Progeny, you would add the table to the Columns pane then check the boxes for each table subfield you wanted on the spreadsheet – in Progeny 9 you merely need to drag the individual table subfield into the Columns pane to add it to the spreadsheet format.

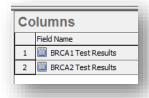
Figure 4-9: The Columns pane of an individual spreadsheet showing table subfields added to it



### Lookup tables in spreadsheets

In Progeny 9, Lookup Table fields are handled the same way as Table fields (see the previous section). Each subfield of the lookup table is displayed in the fields list as a separate entity and you merely need to drag the specific subfield you wish to use in the spreadsheet into the Columns pane.

Figure 4-10: Lookup table subfields added to the Columns pane of a new individual spreadsheet.



# **Entering Data into a Spreadsheet**

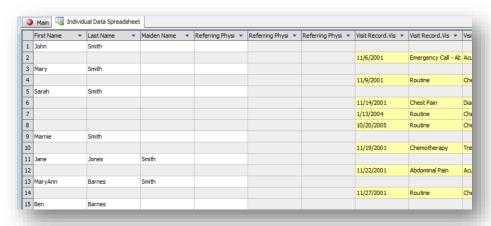
There are a variety of options for entering data into a spreadsheet, including manually <u>entering</u> data, <u>copying</u> and <u>pasting</u> data, <u>finding</u> and <u>replacing</u> data, using the <u>Fill feature</u>, and <u>importing</u> data. Regardless of the method used,

after you have completed data entry in a spreadsheet, click the Save button Save on the spreadsheet toolbar to save the data.

# To manually enter data into a spreadsheet cell

Data entry for the fields in a spreadsheet is very straightforward. Click in the cell and enter the needed data or select the data from a dropdown list. It is possible to enter data into a spreadsheet cell that has no highlighting or one that is highlighted in yellow (a table field). Fields that are highlighted in gray are read-only fields, such as system fields or lookup table subfields, and you cannot enter data directly into these fields. To add more rows to a table, or populate the table subfields for a lookup table, see To add more rows to a table in a spreadsheet below or To add data to a lookup table field in a spreadsheet.

Figure 4-11: Data entry fields and read-only fields in a spreadsheet



To move from one field to the next within the same row, press the Tab key on your keyboard. To move back a field within the same row, press and hold the Shift key and then press the Tab key. To move from one row to the next, press the Enter key. (Pressing the Enter key does not delete the data in a spreadsheet cell.)

#### To add more rows to a table in a spreadsheet

- 1. Select the individual, sample, pedigree, or marker for which you are adding the data, and on the spreadsheet toolbar, click the Add Row button Add Row .
- 2. Click in each table cell of the blank row to add the necessary data.

#### To add data to a lookup table field in a spreadsheet

1. Click on the lookup table field and select the appropriate value for the field.

Figure 4-12: Viewing data in a lookup table in a spreadsheet



- 2. On the spreadsheet toolbar, click the Save button
- 3. On the spreadsheet toolbar, click the Run button Run to populate the table subfields for the lookup table.

### To copy and paste data

It is possible to use standard keyboard commands and/or menu commands to copy text data or numeric data from one cell into another. To copy text in a range of cells, click and hold the left mouse button and drag the mouse to select the region that is to be copied. Once selected, you can copy the data using Edit  $\rightarrow$  Copy, right-click  $\rightarrow$  Copy or CTRL + C on the keyboard. To paste the copied data, select the area where the data will go then use Edit  $\rightarrow$  Paste, right-click  $\rightarrow$  Paste or CTRL + V on the keyboard to paste the data.

## To find and replace data

The Edit menu contains Find and Replace options that you can to search for data in a spreadsheet, and if needed, replace text or numeric data with another value.

Figure 4-13: Find dialog box

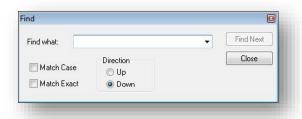


Figure 4-14: Replace dialog box



# To use the Fill feature

The spreadsheet Fill function can fill blank cells with data from adjacent cells if the cells are the same field type. The Fill function is found under the Edit menu and has four options:

- Fill Down fills cells that fall below a selected cell.
- Fill Right fills cells that all to the right of a selected cell.
- Fill Up fills cells that are located above a selected cell.
- Fill Left fills cells that are located to the left of a selected cell.

When filling blank fields that are dropdown list fields, the filled values are stored only in the spreadsheet and not at the database level. For example, the available values for a filled dropdown list field in the spreadsheet are Yes, No, and Pending and you set all the fields to Pending. Although Pending is displayed in these filled fields when editing any of these fields, the only available values will be Received, Due, and N/A, which are the values stored in the database for the field.

#### Figure 4-15: Example of Fill Down/Fill Up

To fill in Amy Brown as the Referring Physician for Patients2 through 12, select the column that includes Amy Brown as the Referring Physician for Patient 1 and the blank Referring Physician cells for Patients 2 through 12, and on the Edit menu, click Fill > Fill Down.

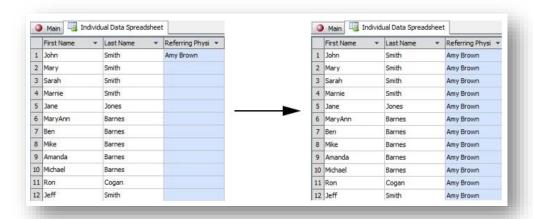
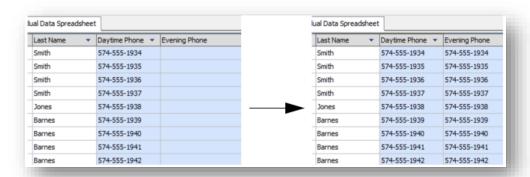


Figure 4-16: Example of Fill Right/Fill Left

To fill in the daytime phone as the evening phone number for Patients 1 through 9 select the column that includes the daytime phone number for Patients 1 through 9 and the column that includes the evening phone number for the same patients, and on the Edit menu, click Fill > Fill Right.

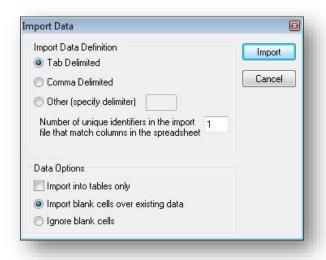


## To import data into a spreadsheet

It is possible to import data into a spreadsheet from a delimited file. The file can be a comma delimited text file, a tab-delimited text file, or you can specify another delimiter type. You can import flat fields (standard fields) or you can import into table rows. If you are importing flat (standard) fields then the data that is in the spreadsheet and the data that you are importing must be the same field type. If importing dropdown fields, and the data that is being imported has different values than the data in the spreadsheet, then the imported values are stored only in the spreadsheet and not at the database level. For example, if the available values for a dropdown list in the spreadsheet are Yes, No, and Pending and the available values for the corresponding dropdown list in the import file are Received, Due, and N/A, and you set the value in the import file to Received, then after the import, "Received" will indeed be displayed in the spreadsheet field; however, when editing the field, the only available values will be Yes, No, and Pending, which are the values stored in the database for the field. If importing data into table rows, then the spreadsheet can contain only the table and its subfields. You can import new rows of table data; however, you cannot import into existing rows of table data.

1. On the spreadsheet toolbar, click the Import button Import. The Import Data dialog box opens. Tab Delimited is selected by default.

Figure 4-17: Import Data dialog box



2. Select the type of delimited file that you are importing. (If the file is not tab delimited or comma delimited, then you must specify the delimiter that the file uses.)

#### 3. Specify the values for the following:

a. The number of Unique Identifiers in the Import File that match columns in the spreadsheet – enter the number of columns in the text file that you are importing that match to the number of columns in the spreadsheet. No data in the matched columns is overwritten. Only data from the matched point forward is overwritten. For example, if your spreadsheet and import text file have three columns each, and you indicate "1" for the number of matched columns, then when the data is imported into the spreadsheet, the data in Column 1 of the spreadsheet is not overwritten; however, the data in Columns 2 and 3 of the spreadsheet is overwritten by the data in Columns 2 and 3 of the import file.



The correct value for the number of matched columns is critical to ensuring the integrity of the data in the spreadsheet

#### b. Data options:

- i. Import into tables only select this option only if you are importing data into table cells in the spreadsheet.
- ii. Import blank cells over existing data allows data in the spreadsheet to be replaced by blank cells.
- iii. Ignore blank cells if the import file contains blank cells, do not replace the data in the spreadsheet with blank cells.
- 4. Click Import. The Open File dialog box opens.
- 5. In the Open File dialog box, browse to select and select the file that you are importing, and then click Open. If there are no errors with the import a message opens indicating the Import operation was successful.
- 6. Click OK. The message closes. You will return to the spreadsheet tab and see the imported information displayed in the spreadsheet.

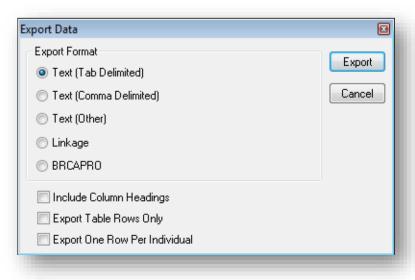
# **Exporting Spreadsheet Data**

Just as you can import data into a spreadsheet from a text file, you can export data from a spreadsheet to a text file. The file can be a comma delimited file, a tab-delimited file, or you can specify another delimiter type. You can export any user-created field as well as system fields.

## To export spreadsheet data

1. On the spreadsheet toolbar, click the Export button Export Data dialog box opens.

Figure 4-18: Export Data dialog box



#### 2. Select the Export format.

- a. If the file is not tab delimited or comma delimited, then you must specify the delimiter that the file uses.
- b. If you select Linkage, then the data is exported to a pedigree file format (.PED). In addition to the exported data, the first five columns of the file also contain the following pedigree relationship fields in the order indicated—UPN, Father ID, Mother ID, and Gender. You must also specify the way in which the individuals in the pedigree are to be identified. The default value is the UPN (Unique Person Number) for each person in the pedigree, but you can select a different value.
- c. If you select BRCAPRO, then the data is exported in a format that can be used in the BRCA Risk Analysis application.

- 3. Select the data that is to be exported.
  - a. Export Table Rows Only—If you are exporting table data and want to export only the table rows that contain data (and not the blank row that indicates a table), then select this option.
  - b. Export One Row per Individual—If you are exporting table data and want to maintain a single row per individual (or sample), then select this option.
- 4. Click Export. The Save As dialog box opens. The Save as Type field is automatically populated with the file type of text (.txt) and you cannot change this value.
- 5. Browse to the location in which the exported file is to be saved, and then in the File name field, enter a name for the file.
- 6. Click Save. A message opens indicating the Export operation was successful.
- 7. Click OK. The message closes. You will then return to the spreadsheet tab.

# Saving and Loading a Spreadsheet Format

After you have generated a spreadsheet with a specific format, you can save the format. You can then retrieve this saved format and load it into another spreadsheet of the same type.



The following procedure details the saving and loading of a spreadsheet format that returns the data for all individuals, pedigrees, samples, or markers in a Progeny database. To save and load a spreadsheet format that returns only a specific subset of data, see Running Spreadsheet Queries

## To save a spreadsheet format



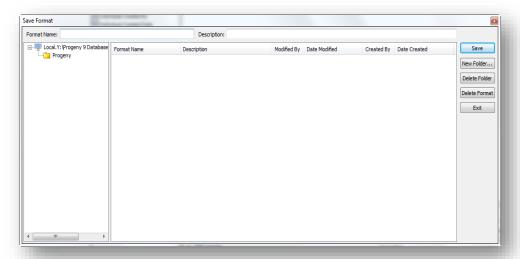
When you save a spreadsheet format, it saves not only the order of the spreadsheet columns but also, the width of the columns.

1. Format and generate the spreadsheet as needed. See Generating a Spreadsheet



2. On the spreadsheet toolbar, click the Save Format Save First button. The Save Format dialog box opens.

Figure 4-19: Save Format dialog box



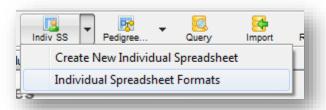
- 3. Enter a name for the spreadsheet format and optionally a description of the spreadsheet format, then do one of the following:
  - a. Select an existing folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the default folders), or it can be a sub-folder of one of the default folders, or of another folder.
    - iii. Click Save.
- 4. Click Save on the Save Format dialog box.

# To load a spreadsheet format

There are two primary methods for loading a saved spreadsheet:

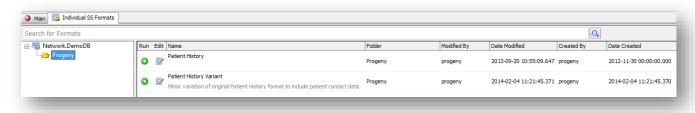
1. Click the drop-down icon next to the Indiv SS button on the main screen and choose Individual Spreadsheet Formats. This opens a new tab with the Individual SS Formats menu.

Figure 4-20: Individual Spreadsheet Formats button on the main menu



- a. Within the Individual SS Formats menu, you can first search for existing formats using the search bar at the top. This is a full-text search across both format names and format descriptions.
- b. The left-side pane will show your spreadsheet format folders while the right-side pane will display a list of formats saved within a selected folder.
  - The right-side pane will display the format name in black text and the description in the grey text. You will additionally be shown the user account that created the format (with timestamp) and the user account that last modified the format (with timestamp)
- c. Click the Run button to launch the spreadsheet or click the Edit button to open the New Individual Spreadsheet menu pre-populated with the selected saved spreadsheet format's parameters.

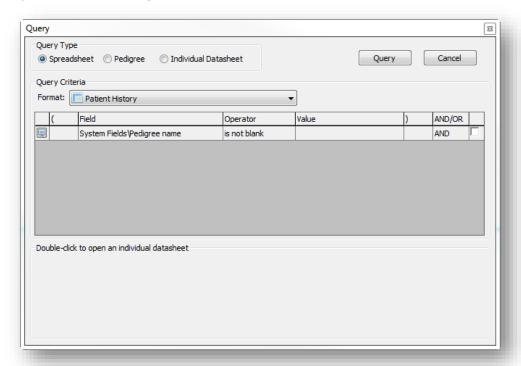
Figure 4-21: Individual Spreadsheet Formats Menu





- 2. Click the Query button Query on the main screen.
  - a. The Query menu offers a shortcut into your saved spreadsheet formats. Choose a saved format from the drop-down menu under Query Criteria and the fields configured for the format's query section will be displayed. You can then make changes to the query parameters.
  - b. Click the Query button on the top right to launch the spreadsheet format with your modified query parameters.

Figure 4-22: Query window activated from the main screen



# **Running Spreadsheet Queries**

You can query the Progeny database to retrieve a specific subset of data. You can run a query one time from within a given spreadsheet, or if you need to run the query frequently, you can save the query as part of the spreadsheet format, and then run the query on an as-needed basis. When you set up the spreadsheet for the query, the spreadsheet does not have to contain the fields against which the query was run. For example, you could set up a query for all female individuals under the age of 50 that have been diagnosed with bilateral breast cancer and the spreadsheet is to display the demographic information for the individuals who meet the query criteria. You would set up the spreadsheet with the following fields—First Name, Last Name, Age at Diagnosis, Current Age, and so on, but you would not need to include the Gender field and the BRCA Diagnosis1 field as these fields would display the same value (Female or BL BR, respectively) for all the individuals who are retrieved by the query.

## To run a spreadsheet query

- 1. <u>Create a new spreadsheet</u> or go into your <u>Individual Spreadsheet Formats</u> and choose the Edit button for your desired spreadsheet format.
  - a. If creating a new spreadsheet add fields into your Columns pane to generate the query output formatting
- 2. Either drag and drop fields from your Fields pane into the Query pane or click the Query button field within your Columns pane.

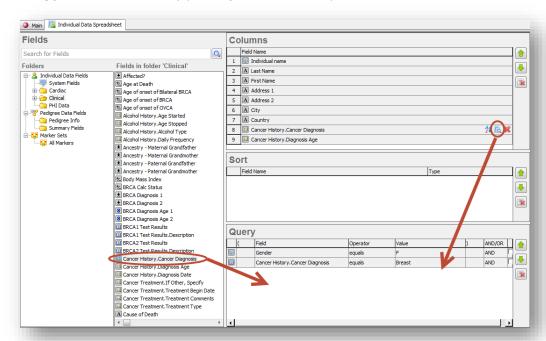


Figure 4-23: Adding fields into the Query pane of an individual spreadsheet

a. If you need to remove a row from your Query pane, either right-click the field and choose Delete Row or click the button on the right side of the query row.



- 3. Click the Run button Bun to run the query. The results of the query are displayed onscreen in the spreadsheet.
  - a. If needed, you can now save this query as a spreadsheet format if you will be using it again in the future (See <u>To Save A Spreadsheet Format</u>)

# Sorting and Filtering Spreadsheet Data

Sorting determines the order of the data that is displayed in a spreadsheet. For example, do you want to display your data based on a birth date that is sorted in reverse chronological order? You can sort the data in a spreadsheet at any time – immediately after you create a spreadsheet, or after you query the spreadsheet data to retrieve a subset of the data. Different sorting options are available for the different types of data columns (text, date, and numeric). After you select a sorting option, the spreadsheet display is dynamically updated.

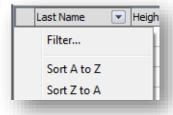
You can also filter spreadsheet data. Filters retrieve a specific subset of the spreadsheet data based on specific constraints (or filters) that you set. For example, you might want to filter spreadsheet data to show only those individuals that were born in the 1970s. You can set multiple filters for a spreadsheet. Multiple filters are mutually inclusive. Only those individuals, pedigrees, samples, or markers that meet all the filtering criteria are displayed in the spreadsheet. As with sorting, you can filter the data in a spreadsheet at any time – immediately after you create a spreadsheet, or after you query the spreadsheet data to retrieve an initial subset of the data.

Although sorting and filtering spreadsheet data are two separate actions, you can combine them as needed to produce a customized dataset that best fits your business needs. For example, you can sort spreadsheet data based reverse chronological birth dates, and you can then filter the sorted data to show only those individuals that were born in the 1970s.

## To sort spreadsheet data

1. Click in the column header for any text, date, or numeric data column. A menu opens with a Filter option and sorting options that are specific to the column type.

Figure 4-28: Example of options for a text column



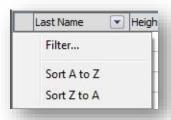
2. Select a sorting option for the column.

Column	Sorting Options and Description
Text	Sort A to Z—Sort the data in the column alphabetical order (from A to Z).
	• Sort Z to A—Sort the data in the column in reverse alphabetical order (from Z to A).
Date	Sort Oldest to Newest—Sort the data in the column in chronological order.
	Sort Newest to Oldest—Sort the data in the column in reverse chronological order.
Numeric	Sort Smallest to Largest—Sort the data in the column in numerical order.
	Sort Largest to Smallest—Sort the data in the column in reverse numerical order.

# To filter spreadsheet data

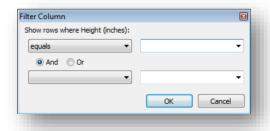
1. Click in the column header for any text, date, or numeric data column. A list opens with a Filter option and sorting options that are specific to the column type.

Figure 4-29: Example of options for a text column



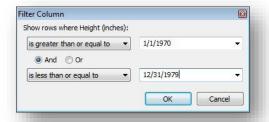
2. Select Filter. The Filter column dialog box opens.

Figure 4-30: Filter Column dialog box



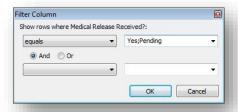
3. Set the filter for the column. For example, Figure 4-31 shows how to filter spreadsheet data to show only those individuals who were born in the 1970s.

Figure 4-31: Filtering for DOB in the 1970s



a. In another example, Figure 4-32 shows how to filter spreadsheet data based on multiple filter values. The values are separated by a semi-colon (;) which acts an OR operator for the filter. (The filter shown here would filter the spreadsheet data based on medical release forms that have been received (Yes) or are pending.)

Figure 4-32: Filtering for release forms with one of two statuses



- 4. Click OK. The spreadsheet display is updated based on the filters that you have set. Any column for which you have set a filter is marked with a Filter icon.
  - a. Optionally, at any time, clear a filter by right-clicking on the column header for the filtered column, and on the context menu that opens, clicking Clear Filter.
- 5. The spreadsheet display is dynamically updated to include all the data in the column.

# **Printing Spreadsheet Data**

After you generate a spreadsheet, you can print the spreadsheet. You can print all the pages in a multipage spreadsheet, or you can print selected pages.

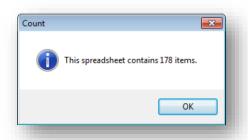
## To print a spreadsheet

- 1. On the spreadsheet toolbar, click the Print button Print . The Print dialog box opens.
- 2. On the Print dialog box, select the needed printing options, and then click Print.

# **Counting Spreadsheet Data**

After you generate a spreadsheet, you can click the Count Button on the spreadsheet toolbar to count the number of individuals, pedigrees, samples, or markers (that is, the number of rows) in the spreadsheet. A Count Dialog box opens, indicating the number of items (rows) that the spreadsheet contains.

Figure 4-33: Count Dialog box



# Chapter 5 Database Security

Database security is the system, processes, and procedures that protect a database from unintended activity be it intentional or unintentional. This chapter details the procedures for setting the appropriate security standards for a Progeny database and its components.

This chapter covers the following topics:

- **Working with Users and Classes**
- **Setting Database Field Security**
- **Setting Database Folder Security**
- **Setting Database Security**
- **Viewing Active Connections**



Except for changing a user password, all functions that are detailed in this chapter are available only to the Progeny administrator. A user can change his/her own password.

# Working with Users and Classes

Progeny always includes an administrator user account by default, called "Progeny". The default administrator password is also "progeny". Only the "Progeny" user has access to the Administration menu and its options for creating, modifying, and deleting other Progeny users and passwords. As the Progeny administrator, after you add a user, you must set the appropriate security levels for the user. The security levels for a user determine the application functions that will be accessible to the user. As the Progeny administrator, you can also add user classes. A user class basically consists of 1 or more of the application functions that you can customize and assign for a user to have. In having a user class, you can control the level of access a user has to all database fields and folders within the database. All users whom you add to the same class will have that same level of access to the exact same database fields and/or folders. As the Progeny administrator, you can also delete users, delete user classes, change the administrator password, and change the passwords for all users.

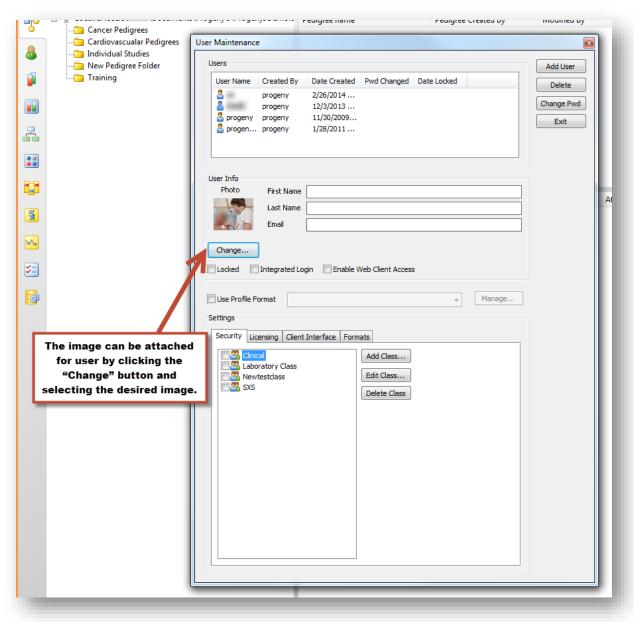


Users can also change their own passwords. See <u>To change your own password as</u> a user

### To add users

1. On the main menu, click Administration  $\rightarrow$  User Maintenance.

Figure 5-1: User Maintenance window





Within the "User Info" pane you can assign the First Name, Last name, Email address and even an image to an existing user. The image can be attached for the user by clicking the "Change" button and selecting the desired image.

2. In the "User Info" pane there are 3 checkbox options:

Option	Description
Enable Web Client Access	To allow the selected user to be able to log in through the web from a browser. (Providing Progeny Web Server is installed).
Integrated Login	This configuration enables the user to log into their Progeny account with authentication from their Windows Profile.
Locked	Enabling this option will lock the selected user's account and prevent them from having any access to the Progeny database.

3. Click Add User. The New User dialog box opens.

Figure 5-2: New User dialog box



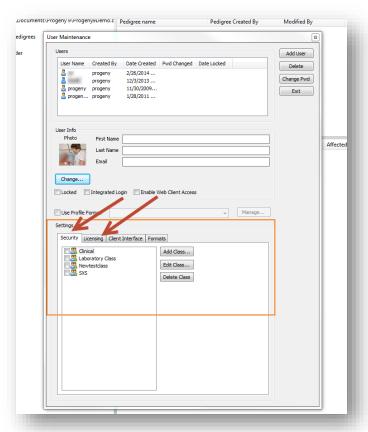
- 4. Enter the username and password for the user account.
  - a. The username and password must adhere to the following rules:
    - i. The username cannot include any spaces.
    - ii. The first character cannot be a number. Instead, it must be an alphabetic character or one of the following special characters: an underscore (\_), an at sign (@), a number sign (#), or a dollar sign (\$).
    - iii. The username can only contain alphabetic characters, numbers, and one or more of the following special characters: an underscore (\_), an at sign (@), a number sign (#), or a dollar sign (\$).

- iv. The username cannot contain a reserved word for an SQL database. See the following:
  - 1. http://www.ianywhere.com/developer/product\_manuals/sqlanywhere/0902/en/html/dbrfen9/0000010.htm
- b. The password is case-sensitive, and it must adhere not only to the same rules as the username, but also, these two rules:
  - i. The password must be a minimum of six characters and contain at least one number.
  - ii. The password cannot contain the username.
- 5. In the "Retype Password" field, enter the password again exactly as you entered it in the Password field.
- 6. Click Add. The Add New User dialog box closes. The new user account is displayed in the Users pane of the User Maintenance window. You can now set security levels for the user account.

## To set the security levels for a user account

- 1. On the main menu, click Administration > User Maintenance to open the User Maintenance window.
- 2. In the Users pane, select the user account for which you are setting the security levels. These security levels can be enabled in the "Settings" pane using two methods: assigning classes or assigning specific and individual functions for the user.

Figure 5-3: User Maintenance Window



- 3. When assigning permissions to users there are two methods for configuring: The Profile Formats and individual assignments using the Security, Licensing, Client Interface and Formats tabs.
  - a. Profile formats allow you to pre-select all options within the four tabs (Security, Licensing, Client Interface and Formats) and tie these to a single format for fast permissions assignment.
    - i. To assign permissions using Profile formats, select the user then check the Use Profile Format box and choose a format from the associated drop-down field.
  - b. To make manual permissions assignments, select a user then work through the four tabs at the bottom of the menu choosing any permissions that you wish to assign. The four tabs are detailed below.

### Security Tab

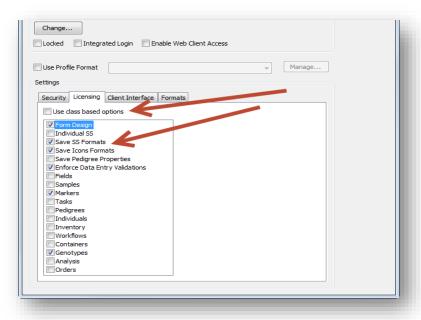
In the security tab, you can assign a user to a user class or create a new user class. To assign a user to a class click on the user then click the checkbox next to the user class you want to assign them to. To create, modify or delete a user class:

- 1. Click "Add Class..." to create a new class, then follow the instructions in the To Add A User Class section.
- 2. Select a user class then click "Edit Class..." and make any desired modifications.
- 3. Select a user class then click "Delete Class..." to remove it.

### Licensing Tab

To assign specific permissions to a user without using "classes" click on the "Licensing" tab within the Settings pane and uncheck the "User class-based options" as seen in the figure below:

Figure 5-4: User Maintenance Window



Select which permissions you want to assign to this user – the table below will explain each of the permissions options.

Option	Description
Form Design	Allow the user to create, edit and add data to datasheets.
Individual SS	Allows the user to create an individual spreadsheet.
Save SS Formats	Allows the user to save a created spreadsheet.
Save Icons Formats	Allows the user to save a created icon format.
Save Pedigree Properties	Allows the user to edit and save options within the Pedigree Properties window.
Enforce Data Entry Validations	Without exception, enforces a rule that this user fills in all fields that are required to have certain values selected. Configured in the "Validations" tab within the "Edit Field" section.
Fields	Allows the user access to the Fields window.
Samples	Allows the user access to the Samples window.
Markers	Allows the user access to the Markers window.
Tasks	Allows the user access to the Tasks window.
Pedigrees	Allows the user access to the Pedigrees window.
Individuals	Allows the user access to the Individuals window.
Inventory	Allows the user access to the Inventory window.
Workflows	Allows the user access to the Workflows window.
Containers	Allows the user access to the Containers window.
Genotypes	Allows the user access to the Genotypes window.
Analysis	Allows the user access to the Analysis window.
Orders	Allows the user access to the Orders window.

### Client Interface Tab

This tab allows you to choose whether the user will access Progeny using the "classic" full interface or the new lightweight "Dashboard" client. The apps listed on this screen allow the admin to grant limited access to specific Progeny functions for a user whom the admin does not want to give full web client access. See <a href="Appendix C">Appendix C</a> for more information on the new Workflows client interface.

#### Formats Tab

The formats tab is an area most users will not need. In this tab, an admin can assign formats to a user – in cases where the folders have default formatting assigned, installing a format into this tab will force Progeny to override the folder configuration and display the user's defaults from this menu instead.

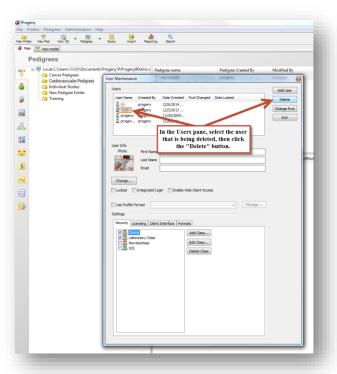
The one extremely useful item here is the Toggle SS format. If configured, when the user clicks the Toggle SS button within the client application Progeny will automatically and immediately display the Toggle SS using the format configured here without prompting or otherwise delaying the user.

### To delete a user

You can delete a user only if the user is not currently logged in to the Progeny database.

- 1. On the main menu, click Administration  $\rightarrow$  User Maintenance. The User Maintenance window opens.
- 2. In the Users pane, select the user that is being deleted.

Figure 5-5: User Maintenance Window



- 3. Click Delete. A message opens asking you if you are sure that you want to delete the selected user.
- 4. Click Yes. A message opens indicating that the user was successfully deleted.
- 5. Click OK to close the message and return to the User Maintenance window.

### To add a user class

A user class contains permissions for the user based on the access that the users are to have to database folders and fields. All users who are in the same class have the same level of access to the same database folders and/or fields. By default, when you add a new user class, the Progeny administrator is automatically added to the class.

- 1. On the main menu, click Administration > User Maintenance. The User Maintenance window opens.
- 2. Within the "Settings" window pane, select the "Security" tab, then click the "Add Class" button. The New User Class window opens.

Figure 5-6: User Maintenance Window

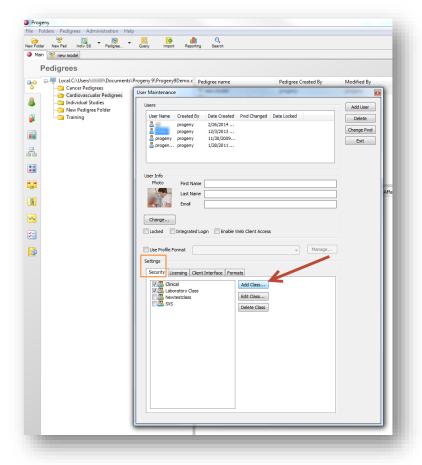
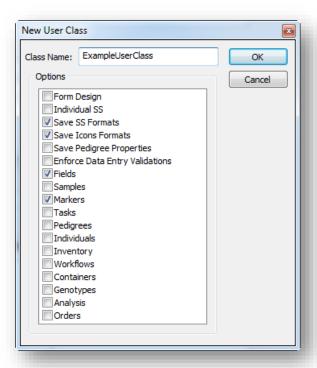


Figure 5-7: New User Class window



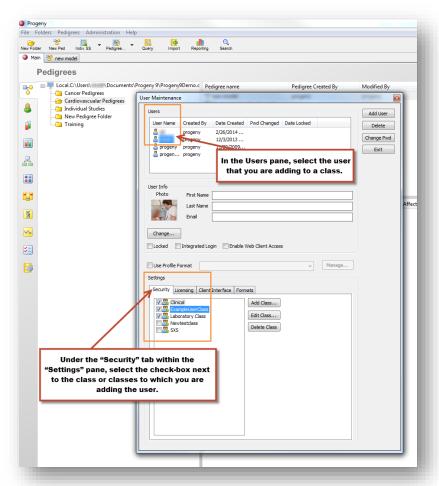
- 3. Enter the name of the new user class, select the desired functions that you would want this class to contain, and then click OK.
  - a. A message opens indicating that the user class was added successfully.
- 4. Click OK to close the message and return to the User Maintenance window.
- 5. You can now add users to the class. See To add users to a user class

### To add users to a user class

By default, as the Progeny administrator, when you add a new user class, you are automatically added to the class. Only non-administrator users must be explicitly added to a user class.

- 1. If you have not already done so, on the main menu, click Administration → User Maintenance to open the User Maintenance window.
- 2. In the Users pane, select the user that you are adding to a class.
- 3. Under the "Security" tab within the "Settings" pane, select the checkbox next to the class or classes to which you are adding the user.

Figure 5-8: User Maintenance Window



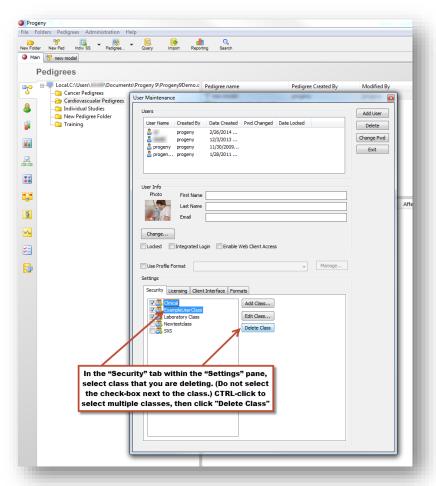
4. Click Exit to close the User Maintenance window. The changes are saved when the window closes. This process can be used to carefully assign each user to their appropriate class. All users who are in the same class have the same level of access to the same database fields and/fields. See <a href="Setting Database Field Security">Setting Database Field Security</a> and <a href="Setting Database Field">Setting Database Folder Security</a>

### To delete a user class

When you delete a class, you delete only the class and not any users who have been assigned to the class; however, all the users who are assigned to the class will lose any security privileges for a database folder and field that was set for that class.

- 1. On the main menu, click Administration  $\rightarrow$  User Maintenance. The User Maintenance window opens.
- 2. In the Users pane, select any user other than the Progeny administrator.
- 3. In the "Security" tab within the "Settings" pane, select the class that you are deleting. (Do not select the checkbox next to the class.) CTRL-click to select multiple classes.

Figure 5-9: User Maintenance Window



- 4. Click Delete. A message opens asking you if you are sure that you want to delete the selected class.
- 5. Click Yes. A message opens indicating that the class was successfully deleted.
- 6. Click OK to close the message and return to the User Maintenance window.

# To change the administrator password

As the Progeny administrator, your default password is progeny. You can change this password, however, if you do change your password, you must record it in a secure location. The password is encrypted and if you lose it or do not remember it, Progeny cannot recover it for you.

1. On the main menu, click Administration → Change Password. The Change Password dialog box opens.

Figure 5-10: Change Password Option

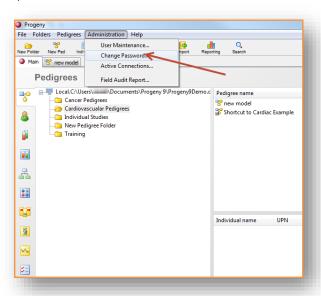


Figure 5-11: Change Password dialog box



- 2. Enter the new administrator password. The new password must adhere to the rules that apply to any user password.
- 3. In the Retype New Password field, enter the password again exactly as you entered it in the New Password field.
- 4. Click OK. The Change Password dialog box closes. You remain logged in to the current Progeny session with your old password. The next time that you log in to Progeny, you must log in using your new password.

# To change a user password as the Progeny administrator



To change the number of days after which all user passwords (other than the administrator password) expire, see <u>Setting Database Security</u>

- 1. On the main menu, click Administration  $\rightarrow$  User Maintenance. The User Maintenance window opens.
- 2. Select the user for whom you are changing the password, then click "Change Pwd" button. The Change Password dialog box opens.

Figure 5-12: User Maintenance Window

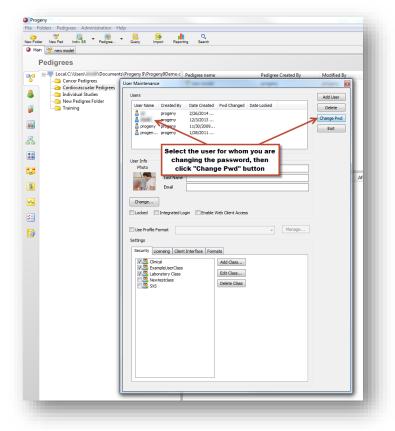


Figure 5-13: Change Password dialog box



- 3. Enter the new user password. The new password must adhere to the rules that apply to any user password.
- 4. In the Retype New Password field, enter the password again exactly as you entered it in the New Password field.
- 5. Click OK. The Change Password dialog box closes. The user remains logged in to the current Progeny session with their old password. The next time that they log in to Progeny, they must log in using their new password.

## To change your own password as a user

1. As a current user, it is possible to change your password once logged into Progeny. On the main menu, click Administration → Change Password. The Change Password dialog box opens.

Figure 5-14: Change Password option

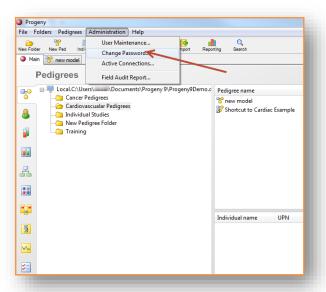


Figure 5-15: Change Password dialog box



- 2. Enter your new user password. The new password must adhere to the rules that apply to any user password.
- 3. In the Retype New Password field, enter the password again exactly as you entered it in the New Password field.
- 4. Click OK. The Change Password dialog box closes. You remain logged in to the current Progeny session with your old password. The next time that you log in to Progeny, you must log in using your new password.

# **Setting Database Field Security**

When you are adding a new database field, then you have the option of setting security for the field. Security determines the type of access (Read, Write or both) that a specific class of user has for the field. The default security settings for any database field are set to Read and Write for All Users. As the Progeny administrator, if you have yet to add any other user classes, then only two default user classes—Clinical and Laboratory—are displayed for a field; otherwise, all the user classes that you have added are displayed. You can add more classes of users if needed.

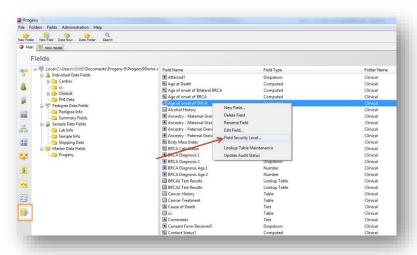


When you create database fields, you typically set the security for the fields on a field by field basis; however, if you want to apply the same security settings for every single field that is contained in the same database folder in a single step, then you can apply the settings at the folder level. See <u>Setting Database Folder</u> Security

# To set database field security

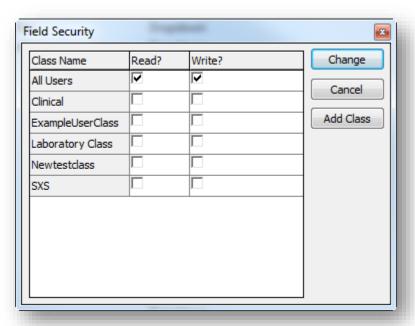
- 1. If the field is already open in the New Data Field dialog box, go to Step 5; otherwise, go to Step 2.
- 2. On the navigation bar, click the Fields button to open the Fields window.
- 3. In the left pane of the Fields window, open the folder that contains the field for which you are setting the security, and then in the right pane of the window, right-click on the field, and on the context menu that opens, click Field Security Level.

Figure 5-16: Field Security Level Option



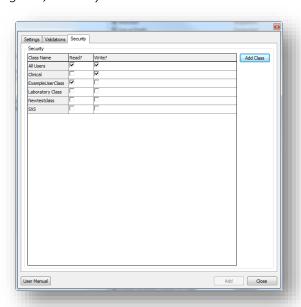
4. The Field Security dialog box opens. Go to Step 6.

Figure 5-17: Field Security dialog box



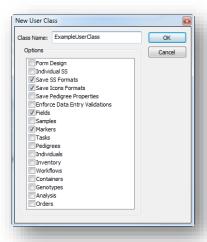
5. Open the Security tab on the New Data Field dialog box

Figure 5-18: Add Data Field dialog box, Security tab



- 6. Do one or more of the following:
  - a. Modify the security for All Users or for the different classes of users.
  - b. Click Add Class to open the New User Class dialog box to create a new user class, and then set the security for the class.

Figure 5-19: New User Class dialog box



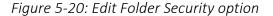
7. Continue to any other necessary tasks for the field or click Add to add the field the database.

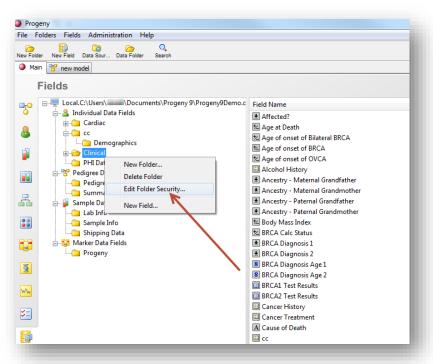
## **Setting Database Folder Security**

When you create database fields, you typically set the security for the fields on a field by field basis; however, if you want to apply the same security settings for every single field that is contained in the same database folder in a single step, then you can set the security at the folder level. The default security settings for any database folder are set to Read Folder and Add/Delete/Modify Fields for All Users. As the Progeny administrator, if you have yet to add any other user classes, then only two default user classes—Clinical and Laboratory—are displayed for a folder; otherwise, all the user classes that you have added are displayed. You can add more classes of users if needed.

#### To set database folder security

- 1. On the navigation bar, click the Fields button to open the Fields window.
- 2. On the Fields window toolbar, right-click on the folder for which you are setting the security, and on the context menu that opens, click Edit Folder Security.



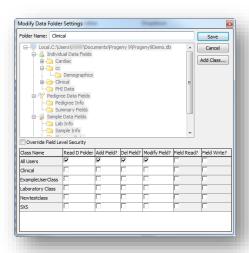


3. The Modify Data Folder Settings dialog box opens. The name of the selected database folder is displayed in the Folder Name field.



Although the dialog box is titled "Modify Data Folder Settings," the selected folder is a database folder.

Figure 5-21: Modify Data Folder Settings dialog box



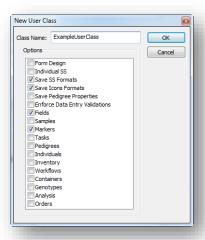
- 4. Do one or more of the following:
  - a. Modify the security settings for All Users or for the different classes of users.
    - i. Read D Folder? —Open and view the contents of the database folder.
    - ii. Add Field? —Add database fields to the folder.
    - iii. Del Field? —Delete database fields from the folder.
    - iv. Modify Field? —Modify database fields in the folder.



To maintain database integrity, you typically do not allow all users the ability to add, delete, and modify fields in a database folder. Instead, only the administrator user has full access to all database folders.

b. Click Add Class to open the New User Class dialog box to create a new user class, and then set the folder security for the new class being added.

Figure 5-22: New User Class dialog box



- i. If you have assigned field-level security to one more fields in the folder, and you want to override the security settings for these fields based on the security settings at the folder level, click Override Field Level Security (Shown in Figure 5-10), and then set the appropriate field-level security for the different classes of users.
  - 1. Field Read? View the data in the field.
  - 2. Field Write? Write data to the field.
- 5. Click Save once security settings have been assigned in "Modify Data Folder" window. A message opens indicating the settings for the folder were successfully changed. Click OK to close the message and return to the Fields window.

## **Setting Database Security**

In addition to setting security at the database field level and the database folder level, you can set security at the database level. When you set security at the database level, you can do one or more of the following:

- Set the number of days after which an inactive user is locked out of the database.
- Set the number of days after which a user's password expires.
- Set the security level for adding, deleting, or modifying database folders for a class of users.
- Set the security level for adding, deleting, or modifying data folders for a class of users, which are the folders that house the data for individuals, pedigrees, samples, and containers.

#### To set database security

- 1. Log in to the database for which you are setting the security.
- 2. On any open window, right-click on the database path, and on the context menu that opens, click Edit Database Security. The Edit Database Security Settings dialog box opens.

Figure 5-23: Database context menu

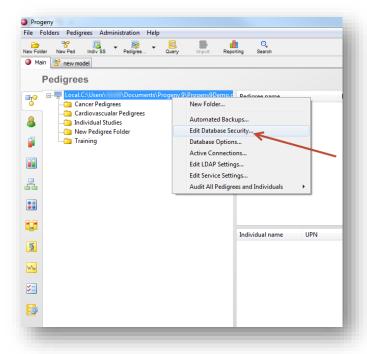
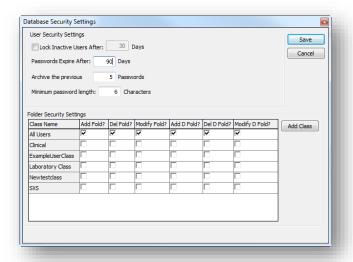


Figure 5-24: Edit Database Security Settings dialog box



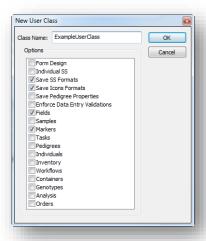
- 3. Set the database security as needed.
  - a. User Security Settings
    - i. Lock Inactive Users After [x] Days Lock out users from the database who have not logged in to the database in the indicated number of days.
    - ii. Passwords Expire After [x] Days The number of days after which all passwords for all users (other than the administrator user) expire. If a user attempts to log in to the database with an expired password, they are prompted to change their password "on the fly" so that they can continue logging in. The password must meet all the requirements for a user password.
    - iii. Archive the previous [x] Passwords Keeps a record of the last 5 passwords held to prevent an unauthorized entry into the database.
    - iv. Folder Security Settings Do one or more of the following:
      - 1. Modify the security settings for All Users or for the different classes of users.
        - a. Add Fold? Add a data folder to the database.
        - b. Del Fold? Delete a data folder from the database.
        - c. Modify Fold? Modify a data folder in the database.
        - d. Add D Fold? Add a database folder to the database.
        - e. Del D Fold? Delete a database folder from the database.
        - f. Modify D Fold? Modify a database folder in the database.



To maintain database integrity, you typically do not allow all users the ability to add, delete, and modify fields in a database folder.

2. Click Add Class to open the New User Class dialog box to create a new user class, and then set the folder security for the new class.

Figure 5-25: New User Class dialog box



4. On the "Database Security Settings" menu, click Save. A message opens indicating the settings for the database were successfully changed. Click OK to close the message and return to the application.

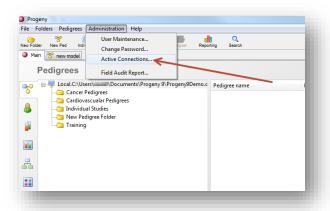
### **Viewing Active Connections**

As the Progeny administrator, you can view all active connections to any Progeny database. You typically view active connections for security purposes, for example, to ensure that the database is not being improperly accessed by users. If you determine that a user who is accessing the database should not be, you can edit the database security settings to immediately lock the user out of the database.

#### To view active connections

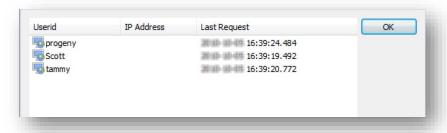
1. Log in to the database for which you are viewing the connections, then click Administration → Active Connections.

Figure 5-26: Active Connections Option



The Active Connections window opens. This window lists all active connections by username and IP address. It also lists a date and timestamp of the last login request for the user.

Figure 5-27: Active Connections window



Optionally, if you determine that a user who is accessing the database should not be, you can edit the database security settings to immediately lock the user out of the database. See <u>To set the security levels for a user account</u>.

# Chapter 6 – Database Reporting

Progeny contains a general reporting function to assist you in monitoring and maintaining your database records. You can generate reports for all individuals, pedigrees, markers, and samples in a Progeny database. The reporting function also contains a Query option which you can use to define custom criteria for searching for a specific subset of data in a Progeny database.

This chapter covers the following topics:

- Generating a Report
- Saving and Loading Report Formats

# Generating a Report

Progeny contains a general reporting function to assist you in monitoring and maintaining your database records. You can generate reports for individuals, pedigrees, samples, and markers.



The following procedure details the generation of a Sample report, but, by analogy, you can apply this procedure to generate reports for individuals, pedigrees, and markers.

### To generate a report

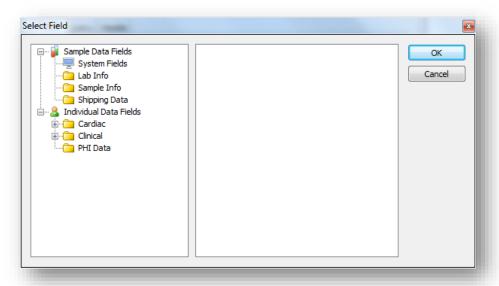
- 1. On the Progeny main window navigation bar, click the Samples button to open the Samples window.
- 2. On the window toolbar, click the Reporting button Reporting.
  - a. The Report Setup dialog box opens. The Fields tab is the active tab.

Figure 6-1: Report Setup dialog box



- 3. Right-click on the Report Setup dialog box, and on the context menu that opens, click Add Field.
  - a. The Select Field dialog box opens.

Figure 6-2: Select Field dialog box



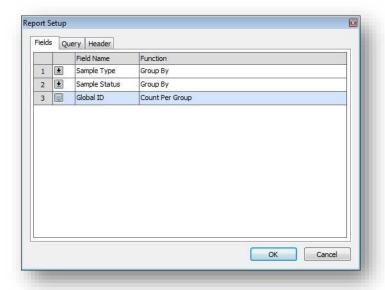
- 4. Select a field from the Select Field dialog box and then click OK.
  - a. The Select Field dialog box closes, and the field is displayed on the Fields Tab of the Report Setup dialog box.
- 5. Repeat Step 3 and Step 4 for each field that is to be displayed in the report.



The order in which you add the fields to the Report Setup dialog box is the order in which they appear (from left to right) as columns in the report. To delete a field from the report, or to change a field section, right-click on the appropriate field, and on the context menu that opens, click Delete Field or Change Field, respectively.

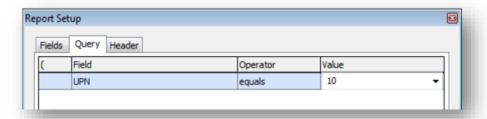
- 6. For each report field, select a function.
  - a. The default value is Grouped By. The report output is grouped according to the functions that you select from top to bottom. For example, in Figure 6-3 below, the report is first grouped by Sample Type, and then within each sample type, the report is grouped by Sample Status. If your database contains five sample types and six sample statuses, then your report could contain up to a maximum of 30 rows. If you select one of the Per Group functions (Count Per Group, Max Per Group, or Min Per Group), then for optimum results, this function should be the last function that is selected, and you must select a system field that is relevant for the item that you are counting. (Again, see Figure 6-3 below.) For example, if you are counting samples, then you could select the Barcode system field to count the total number of samples in the database or you could select Global ID to count all the samples in the database that are linked to an individual.

Figure 6-3: Example of a report setup



- 7. You can optionally open the Query tab and define a query to further filter the report results or click OK on the Report Setup dialog box to generate the report.
  - a. Open the Query tab and define a query to further filter the report results.
    - i. For example, to filter the report described in Figure 6-3 above by samples that are linked to a specific individual, you would set up the query as shown in Figure 6-4 below.

Figure 6-4: Report Setup. An example guery





See <u>Database Query Format</u> for more detailed information about formatting a database query.

- b. Open the Header tab and enter a header that is displayed at the top of every page in the report.
- 8. Click OK. The Report Setup dialog box closes, and the report is generated and displayed onscreen. Figure 6-5 below shows the Sample report that is generated without the query (675 total samples in the database are linked to an individual). Figure 6-6 below shows the Sample report that is generated by the query (Of these 675 samples, 43 are linked to the individual whose UPN = 10).

Figure 6-5: Sample report, no query

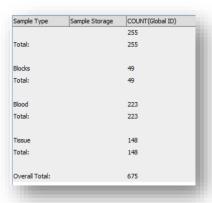
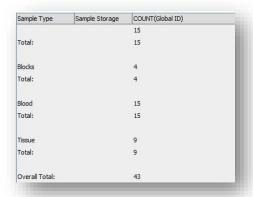


Figure 6-6: Sample report, with query



- 9. Optionally, after generating a report you can do one or more of the following:
- 10. Click the Setup button Setup to open the Report Setup dialog box, modify the report settings, and generate the report again.
- 11. Click the Print button Print to print the report.
- 12. Click the Export button Export to export the report. See Exporting Spreadsheet Data
- 13. Save the report format for loading and running as required.

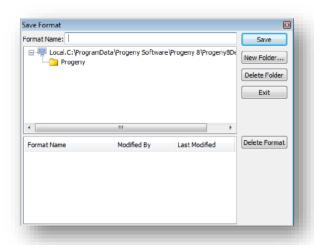
## Saving and Loading Report Formats

After you have created a report with a specific format, you can save the format. You can then load this saved format and use it to generate another report of the same type.

#### To save a report format

- 1. Format and generate the report as needed. See Generating a Report
- 2. On the report toolbar, click the Save Format button Save Fmt .

Figure 6-7: Save Format dialog box



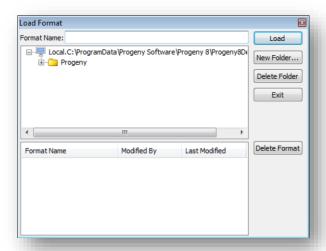
- 3. Enter a name for the report format, and then do the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
    - iii. Click Save.
- 4. Click Save on the Save Format dialog box.

#### To load a report format

You can a retrieve a saved report format and use it to generate a report of the same type.

- 1. On the Progeny main window navigation bar, open the window for the item for which you are generating the report. For example, if you are generating a Sample report, click the Samples button to open the Samples window.
- 2. On the window toolbar, click the Reporting button
- 3. On the window toolbar, click the Load Format button Load Fmt
  - a. The Load Format dialog box opens.

Figure 6-8: Load Format dialog box



- 4. Open the folder that contains the report format that you are loading, select the format, and then click Load.
  - a. The Report Setup dialog box opens. The dialog box displays the information for the loaded format (selected fields, functions etc.).
- 5. Click OK on the Report Setup dialog box.
  - a. The report is generated and displayed onscreen according to the loaded format

# Chapter 7 Database Organization

Progeny provides two functions—Tasks and Smart Lists—to assist you in managing a Progeny database and organizing the data for easy review and retrieval.

This chapter covers the following topics:

- <u>Using the Tasks Feature</u>
- Creating Smart Lists

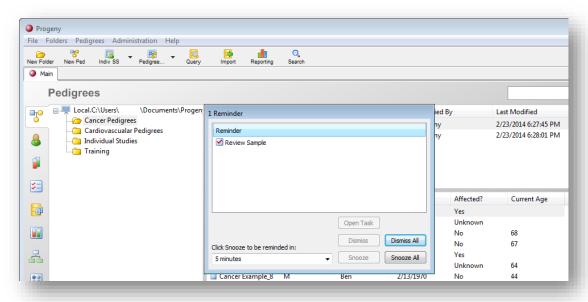
### Using the Tasks Feature

The Tasks feature centralizes the database tasks that have been created by you or assigned to you in a single location. After you log into Progeny, if any tasks that are assigned to you have been flagged with a reminder that has a due date less than or equal to the current day's date, then a Reminders dialog box opens in the Tasks main window. The dialog box lists the tasks by name. You can create tasks in the Tasks window and assign the task to yourself or to another user, and you can also edit and delete these tasks. If a task has not been created with a reminder, or if the current date is too soon for a reminder and therefore, the task is not displayed in the Reminders dialog box, you can search for the task in the Tasks window.

#### To work with tasks in the Reminders dialog box

After you log into Progeny, if any tasks that are assigned to you have been flagged with a reminder that has a due date less than or equal to the current day's date, then a Reminders dialog box opens in the Tasks main window. The dialog box lists the tasks by name.

Figure 7-1: Tasks window with Reminders dialog box



- To view a task in the Reminders dialog box, select the task and then click Open Task.
- To dismiss a task from the Reminders dialog box (regardless of its status), select the task and click Dismiss.
  - o To dismiss all tasks from the Reminders dialog box, click Dismiss All.



When you dismiss a task from the Reminders dialog box, the task is not deleted. A dismissed task can still be returned in a task search. If you dismiss all tasks, the Reminders dialog box closes.

- To change the default task reminder Snooze time from 5 minutes to another time for a single task, select the task, select a different time on the Snooze time dropdown list, and then click Snooze.
- To change the default task reminder Snooze time from 5 minutes to another time for all tasks, select a different time on the Snooze time dropdown list, and then click Snooze All.



If you snooze all reminders, the Reminders dialog box closes.

#### To create a database task

You can create a database task from the Tasks window, or you can manually associate an item (individual, sample, or pedigree) with a task. If you create a task from the Tasks window, then you must associate an automatic query with the task. When the user selects the task in the Tasks window, the associated query is automatically generated, and the results of the query are displayed in the lower right pane of the Tasks window.

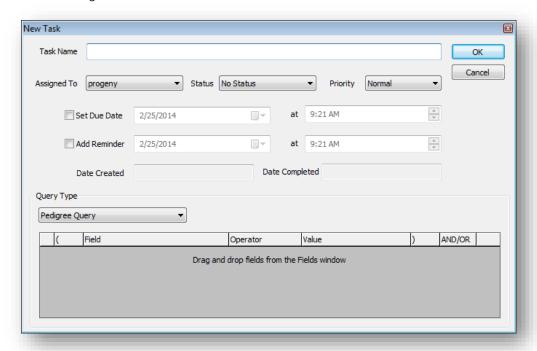


Make sure that you are logged in to the Progeny database for which you want to create the task.

#### To create a task from the Task window

- 1. On the Progeny main window, click the Tasks button to open the Tasks window.
- 2. On the Tasks window toolbar, click the New Task button New Task . The New Task dialog box opens.

Figure 7-2: New Task dialog box

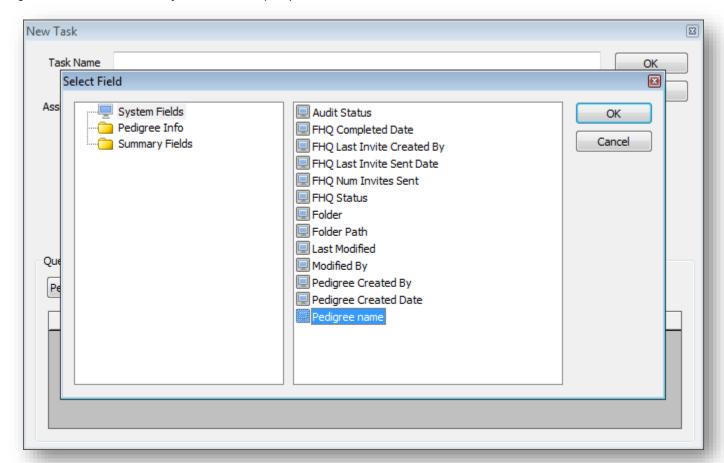


3. Enter the identifying information for the task.

Field	Description
Task Name	A descriptive or identifying name for the task.
Assigned To	A dropdown list of all the users in the database.
Status	Dropdown list. Available values are No Status, In Progress, and Completed.
Priority	Dropdown list. Available values are Normal, Low, and High.
Set Due Date Add Reminder	Default values are the current day's date and the current time. To modify these values, select the appropriate checkbox to enable the Date and Time fields.  Note: If you do not set a reminder for a task, then to view the task, you or the user to whom you assigned the task must search for the Task in the Tasks window.

4. To associate an automatic query with the task, select the Query Type (Pedigree Query, Individual Query, or Sample Query), right-click in the Query pane to add a row, then select a field from a fields folder.

Figure 7-3: Field selection for New Task query





See <u>Database Query Format</u> for detailed information about formatting a database query.

- 5. Choose the field you want to query on and click OK.
- 6. The Add New Task dialog box closes. The newly created task is displayed in the upper right pane of the Tasks window.

#### To manually associate an item with a task

1. Right-click on the item (individual, pedigree, or sample) that you are associating with a task, and on the context menu that opens, click New Task. The New Task dialog box opens, and you will see the selected item displayed at the bottom under Associated Items.

Figure 7-4: Creating a task for an individual

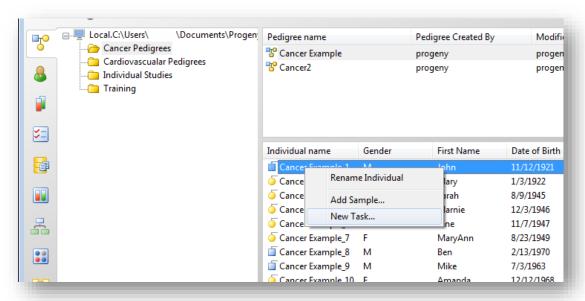
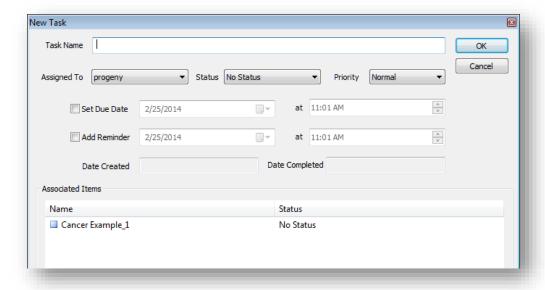


Figure 7-5: New Task dialog box



2. Enter the information for the task.

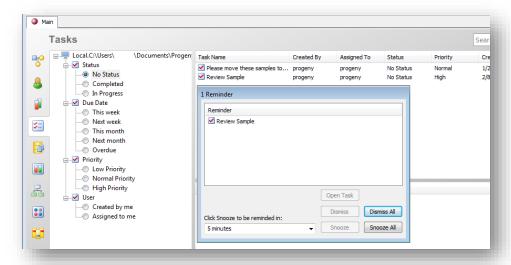
Field	Description
Task Name	A descriptive or identifying name for the task.
Assigned To	A dropdown list of all the users for the database.
Status	Dropdown list. Available values are No Status, In Progress, and Completed.
Priority	Dropdown list. Available values are Normal, Low, and High.
Set Due Date Add Reminder	Default values are the current day's date and the current time. To modify these values, select the appropriate checkbox to enable the Date and Time fields.
	Note: If you do not set a reminder for a task, then to view the task, you or the user to whom you assigned the task must search for the Task in the Tasks window.

3. Click OK. The Add New Task dialog box closes. The newly created task is displayed in the upper right pane of the Tasks window.

#### To edit a task

- 1. Log in to the Progeny database in for which you are editing the tasks.
  - a. If any tasks that are assigned to you have been flagged with a reminder that has a due date less than or equal to the current day's date, then a Reminder dialog box opens in the Tasks main window. The dialog box lists the tasks by name.

Figure 7-6: Tasks window with Reminder dialog box



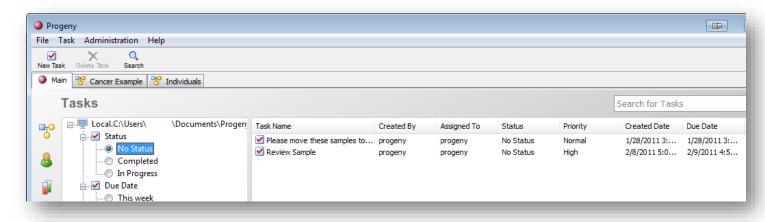
#### 2. Do one or more of the following:

- a. If the task that you are editing is displayed in the Reminders dialog box, select the task, and then click Open Task. The Tasks opens in the Edit Task dialog box. Edit the task as needed.
- b. If the task that you are editing is not displayed in the Reminders dialog box, then do one of the following:
- c. Carry out a contextual search for the task. See Contextual Search.
- d. Select the appropriate option in the left pane of the Tasks window to search by Status, Due Date, Priority, or User.



The search options in the left pane are mutually exclusive. You cannot, for example, search for all tasks that you created that have a high priority. If you search for all tasks that you created, then all tasks that you created are returned regardless of their priority. The tasks the meet the search criteria are displayed in the upper right pane of the Tasks window.

Figure 7-6: Tasks search results in Tasks window

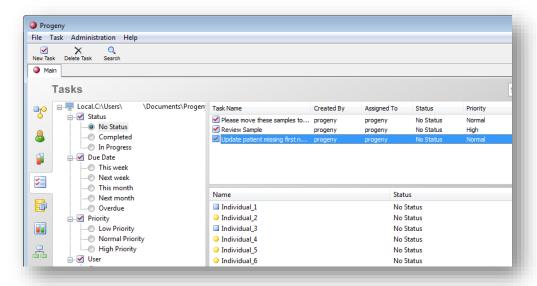


3. Double-click on the task in the Tasks window to open the Tasks in the Edit Tasks window and edit the task as needed.



If an automatic query is associated with the selected task, then the results of the query are displayed in the lower pane of the Tasks window.

Figure 7-7: Query results for an automatic query associated with a task



#### To delete a task

- 1. Log in to the Progeny database for which you are deleting the tasks.
- 2. If applicable, close the Reminders dialog box, Snooze All tasks or Dismiss All Tasks.



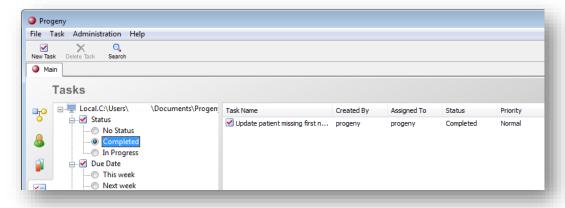
It is recommended to Snooze All Tasks to close the dialog box as Dismissing All Tasks will permanently remove the tasks from the Reminders list.

- 3. Do one of the following:
  - a. Carry out a contextual search for the task. See **Contextual Search**.
  - b. Select the appropriate option in the left pane of the Tasks window to search by Status, Due Date, Priority, or User.



The search options in the left pane are mutually exclusive. You cannot, for example, search for all tasks that you created that have a high priority. If you search for all tasks that you created, then all tasks that you created are returned regardless of their priority.

Figure 7-8: Tasks search results in Tasks window



- 4. Select the task or tasks that are being deleted, (CTRL-click to select multiple tasks), and then on the Tasks window toolbar, click the Delete Task button.
- 5. A message opens asking you if you are sure that you want to delete the selected tasks.
- 6. Click Yes. The message closes and the selected tasks are deleted.

### To search for a task

You have two options for searching for a task from the Tasks window:

- 1. Do a contextual search for the task. See <u>Contextual Search</u>.
- 2. Click the Search icon Search on the Tasks window toolbar to carry out a global search. See <u>Database Global Search</u>.

### **Creating Smart Lists**

When you collect data for an individual, pedigree, marker, and so on, you typically have multiple fields in which to enter the data. For example, if you are collecting data for an individual, the fields in which you must enter data can include First Name, Last Name, Maiden Name, Address 1, Address 2, Address 3, City, State, Zip, Race, DOB, Current Age, Height, Weight, etc. When you are trying to quickly locate a specific individual, pedigree, etc....in a data folder, it is simply not feasible to display all the fields. Instead, you can customize the fields that are displayed in a smart list. A smart list displays only selected fields from all the available data fields and system fields for individuals, pedigrees, samples, markers, and inventories. Smart lists are displayed in the right panes of the Progeny main window. Multiple smarts lists are available for individuals, pedigrees, samples, and inventories. Only a single smart list is available for markers. (See Smart List types). A smart list can be saved on a per-user basis so that each user can have his/her own preferred view, or the Progeny administrator can set a single default view for all users. After you or the Progeny Administrator creates the smart lists of individuals, pedigrees, etc., you can carry out a contextual search. A contextual search searches only the data that is displayed in the columns in a smart list. This provides a convenient method for quickly retrieving needed data. (See Contextual Search.)

Figure 7-9: Individual Smart List example

Pedigree name		Pedigree Created By	Modified	Ву	Last Modified
Cancer Example		progeny	progeny		2/23/2014 6:27:45 PM
<mark>™</mark> Cancer2		progeny	progeny		2/23/2014 6:28:01 PM
Individual name	Gender	First Name	Date of Birth	Affected?	Current Age
Cancer Example_1	М	John	11/12/1921	Yes	
Cancer Example_1			,,	103	
Cancer Example_2	F	Mary	1/3/1922	Unknown	
	F F	Mary Sarah			68
Cancer Example_2		•	1/3/1922	Unknown	68 67
Cancer Example_2 Cancer Example_4	F	Sarah	1/3/1922 8/9/1945	Unknown No	-
Cancer Example_2 Cancer Example_4 Cancer Example_5	F F	Sarah Marnie	1/3/1922 8/9/1945 12/3/1946	Unknown No No	-

### Smart List types

You can define smart lists for pedigrees, individuals, samples, aliquots, markers, tasks, inventory and containers.

Figure 7-10: Pedigree and Individual smart lists

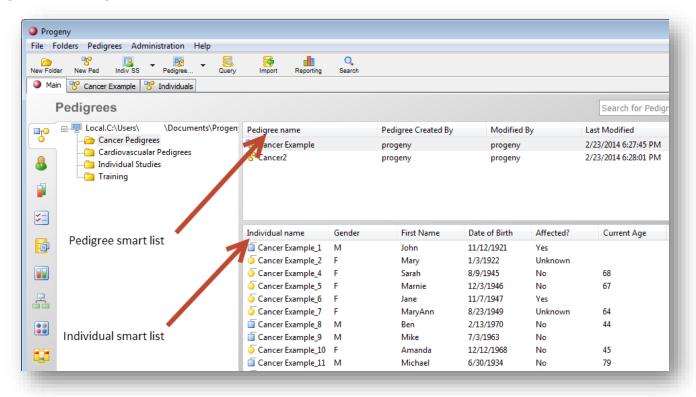
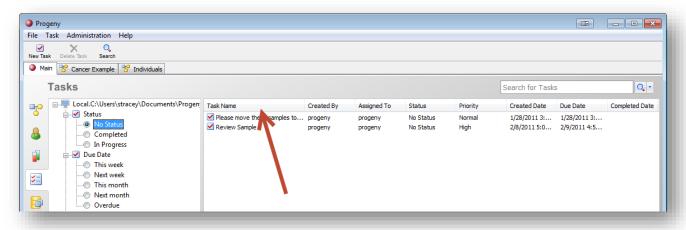


Figure 7-11: Tasks smart list



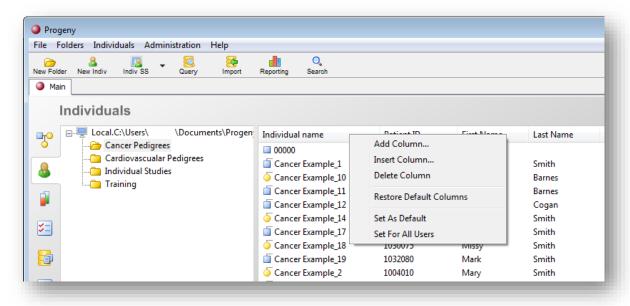
#### To create a smart list



The following procedure details the creation of an individual smart list, but, by analogy, you can apply this procedure to create smart lists for pedigrees, samples, inventories, and markers.

- 1. Click the Individuals button bt to open the Individuals window.
- 2. Right-click in any of the column headers for a smart list. The smart list context menu opens.

Figure 7-12: Smart List context menu



3. Select the appropriate option on the context menu.

Option	Description
Add Column	Add a new column at the end of the smart list.
Insert Column	Add a new column to the immediate left of the selected column.
Delete Column	Delete the selected column from the smart list.
Restore Default Columns	Return the smart list to its default view. This view can be the default installation view, or it can be a default view that the Progeny administrator has set for all users.
Set as Default	Available only for the Progeny administrator. Sets the current view as the default view for the smart list.
Set for All Users	Available only for the Progeny administrator. Sets the current view as the default view for the smart list for all progeny users.



Remember, after you or the Progeny Administrator creates smart lists for individuals, pedigrees, and so on, you can carry out a contextual search. A contextual search searches only the data that is displayed in the columns in a smart list, which provides a convenient method for quickly retrieving needed data. See Contextual Search.

# Chapter 8 Database Queries and Searches

Both queries and searches are an inquiry into the database and both are used to extract data from the database in a readable format according to a user's request. Queries and searches differ primarily in the operators that are used in the request and the allowed format of the request.

This chapter covers the following topics:

- Database Query Format
- Database Global Search
- Contextual Search
- Main Window Queries

# **Database Query Format**

A database query can be used to extract data from the database in a readable format according to a user's request. A query can be carried out for a single field in the database, or it can be carried out for multiple fields. For either type of query, after you have selected the fields for the query, you must:

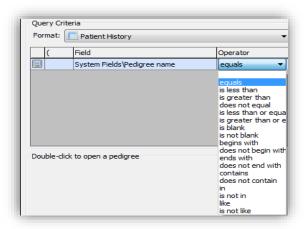
- Specify the operators for the query.
- Specify the values for the query. Depending on the fields that you selected for the query, you must either manually enter a value, or select a value from a table or dropdown list.

In addition, if you are carrying out a multiple fields query, you must specify the <u>Boolean operators</u> (AND/OR) for the query. Optionally, you can also use brackets to apply an <u>order of operations</u> to the query, which determines the order the queries are carried out.

#### Query operators

When you are setting up a database query, you must select from a list of operators for the query.

Figure 8-1: List of available operators for a query



Operator	Description
equals	Search for an exact match. For example, if the query field for individuals is the Weight field, and you specify the search criteria as Weight = 175, then only those individuals whose weight is exactly 175 are returned in the search.
is less than	Search for an item that is less than the specified value. For example, if the query field for individuals is the Weight field, and you specify Weight < 175, then only those individuals whose weight are less than 175 are returned in the search.
is greater than	Search for an item that is greater than the specified value. For example, if the query field for individuals is the Weight field, and you specify Weight > 175, then only those individuals whose weight is greater than 175 are returned in the search.
does not equal	Search for an item that meets any criteria other than the specified value, for example, if the query field for individuals is the Weight field, and you specify Weight ≠ 175, then only those individuals whose weight does not equal 175
is less than or equal to	Search for an item that is less than the specified value or is an exact match for the specified value. For example, if the query field for individuals is the Weight field, and you specify Weight ≤ 175, then those individuals whose weight is exactly 175 or less are returned in the search.
is greater than or equal to	Search for an item that is greater than the specified value or is an exact match for the specified value. For example, if the query field for individuals is the Weight field, and you specify Weight ≥ 175, then those individuals whose weight is exactly 175 or greater are returned in the search.
is blank	Search for an item where the specified field contains absolutely no data. For example, if the query field for individuals is the Weight field, and you specify Weight is blank, only those individuals for whom data has not been entered, the Weight field are returned in the search.  Note: A zero (0) is an actual data value. This means that if a zero (0) has been entered, the Weight
	field for an individual, then the individual is not returned in the search.
is not blank	Search for an item where the specified field contains any valid data. For example, if the query field for individuals is the Weight field, and you specify Weight is not blank, all individuals for whom data has been entered, the Weight field are returned in the search.
	Note: A zero (0) is an actual data value. This means that if a zero (0) has been entered, the Weight field for an individual, then the individual is returned in the search.
begins with	Search for an item where the specified field contains data that begins with the search criteria. For example, if the query field for individuals is the Weight field, and you specify Weight begins with 17, then only those individuals for whom the weight begins with a 17 (17 or 170-179) are returned in the search.
does not begin with	Search for an item where the specified field contains data that does not begin with the search criteria. For example, if the query field for individuals is the Weight field, and you specify Weight does not begin with 17, then only those individuals for whom the weight does not begin with a 17 (0-16, 18 -169, and so on) are returned in the search.

ends with	Search for an item where the specified field contains data that ends with the search criteria. For example, if the query field for individuals is the Weight field, and you specify Weight ends with 75, then only those individuals for whom the weight ends with a 75 (75, 175, 275, and so on) are returned in the search.
does not end with	Search for an item where the specified field contains data that does not end with the search criteria. For example, if the query field for individuals is the Weight field, and you specify Weight does not end with 75, then only those individuals for whom the weight does not end with a 75 (0-74, 76-174, 176-274, and so on) are returned in the search.
contains	Search for an item where the specified field contains an instance of the search criteria. For example, if the query field for individuals is the Weight field, and you specify Weight contains 75, then all individuals who have a weight in which 75 appears anywhere (75, 175, 275, and so on) are returned in the search.
does not contain	Search for an item where the specified field does not contain any instance of the search criteria. For example, if the query field for individuals is the Weight field, and you specify Weight does not contain 75, then all individuals who have a weight in which 75 does not appear (0-74, 76-174, 176-274, and so on) are returned in the search.
in	Allows for comma separated values to be entered in a field so that multiple items can be searched and returned in the search results. For example, if the query field for individuals is the Weight field, and you specify Weight in 75, 80, 85, 90, then all individuals whose weight is exactly 75, 80, 85, or 90 are returned in the search.  Note: You can copy and paste a list of values from a .csv file into a query of this type.
is not in	Allows for comma separated values to be entered in a field so that multiple items can be searched and excluded from the search results. For example, if the query field for individuals is the Weight field, and you specify Weight is not in 75, 80, 85, 90, then all individuals whose weight is not exactly 75, 80, 85, or 90 are returned in the search.  Note: You can copy and paste a list of values from a .csv file into a query of this type.
like	A pattern matching search based on the keyword "like." An underscore (_) is used to match exactly one character, and the percent sign (%) is used to indicate any number of characters. For example, if the query field for individuals is the Weight field, then to search for all individuals whose weight is like 75, select "like" as the operator and enter %75 in the Value field. The % indicates that any number of characters can come before 75, for example, 75, 175, 275, and so on.  Note: You can use the % anywhere in the search criteria, for example, 75% or 7%5.
is not like	A pattern matching search based on the keyword "like." An underscore (_) is used to match exactly one character, and the percent sign (%) is used to indicate any number of characters. For example, if the query field for individuals is the Weight field, then to search for all individuals whose weight is not like 75, select "is not like" as the operator and enter %75 in the Value field. The % indicates that any number of characters can come before 75, for example, 0 -74, 176 - 184, 186 - 274, and so on.  Note: You can use the % anywhere in the search criteria, for example, 75% or 7%5.

#### AND/OR operators

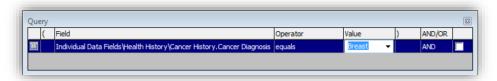
You use the Boolean operators AND and OR to narrow or widen your search respectively.

A search with the Boolean operator AND returns only those items that use both the search terms you specify, as in this example: BREAST AND FEMALE.

A search with the Boolean operator OR returns those items that use either search term that you specify, as in this example: BREAST OR FEMALE.

When you set up a single field query, then by default, the AND/OR operator is set to AND, which is ignored by the application. For example, Figure 8-2 shows the query for all individuals with a cancer diagnosis equal to Breast.

Figure 8-2: Singe field query



When you set up a multiple fields query, then you must specify the correct operator for the desired query results. For example, Figure 8-3 shows the query for all individuals who have a cancer diagnosis equal to Breast or who were diagnosed with any type of cancer at the age of 65.

Figure 8-3: Multi-field query with OR operator

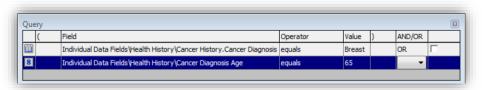
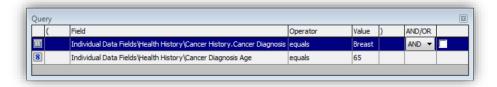


Figure 8-4 shows the query for all individuals who have a cancer diagnosis equal to Breast and who were diagnosed with this type of cancer at the age of 65.

Figure 8-4: Multi-field query with AND operator



#### Order of operations for a query

When you carry out a multiple fields query, you can use parentheses to define the order of operations for a query. The order of operations determines the queries that must be carried out first in a series of queries. For example, Figure 8-5 shows a query that first sorts individuals who were diagnosed with breast cancer at the age of 65. The query then sorts individuals who were diagnosed with colon cancer. Because this query uses the OR operator, the query returns individuals who meet either query criteria.

Figure 8-5: Order of operations for the multi-field query, OR operator

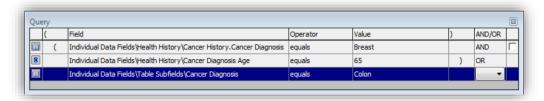
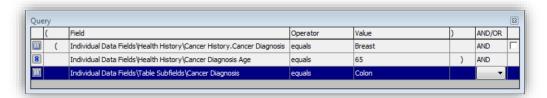


Figure 8-6 shows a query that first sorts individuals who were diagnosed with breast cancer at the age of 65. The query then sorts individuals who were diagnosed with colon cancer. Because this query uses the AND operator, the query returns only those individuals who meet both query criteria.

Figure 8-6: Order of operations for the multi-field query, AND operator



#### **Database Global Search**

When you carry out a database global search, the entire database is searched for every occurrence of a sequence of characters, or search string. Your search is limited to the exact order of the characters in the string and you can use the asterisk (\*) as a wildcard to extend the search. For example, if you are searching for individuals with some form of Bob in either their First Name or Last Name:

If you enter Bob as the search string, then only those database items that contain the exact value of "Bob" in at least one of the data fields are returned by the search, for example, the individual Bob Smith.

If you enter Bob\* as the search string, then the database items that contain the value "Bob" followed by any other characters in at least one of the data fields are returned by the search, for example, the individual Bobbi Smith or the individual James Bobby.

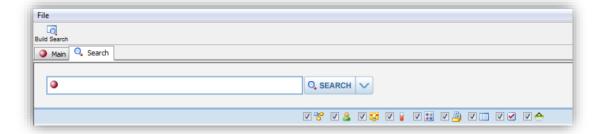
If you enter \*Bob as the search string, then the database items that contain the value "Bob" preceded by any other characters in at least one of the data fields are returned by the search, for example, the individual Jim-Bob Walton.

All items that are returned by the search are returned in a hyperlink format, which you can click to go to the referenced item.

#### To carry out a database global search

- 1. On the Progeny main window toolbar, click the Search icon. Search.
- 2. A blank Search tab opens. By default, all database items for which you can search (from left to right, the icons are for pedigrees, individuals, markers, samples, containers, fields, spreadsheets, tasks, and orders) are selected.

Figure 8-7: Search tab with default search options



- 3. Optionally, clear the selections for the database items for which you do not want to search.
- 4. In the Search field, enter the search string. Use asterisks as needed to extend the search.
- 5. Click Search. All database items that meet the search criteria are returned by the search. The items are returned in a hyperlink format, which you can click to go to the referenced item.

### Contextual Search

When you carry out a contextual search, only the data that is displayed in the columns in a smart list is searched. Your search is limited to the exact order of the characters in the string and you can use the asterisk (\*) as a wildcard to extend the search. For example, when searching an individual smart list where the First Name is one of the columns that is displayed:

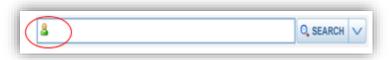
If you enter Bob as the search string, then only those individuals that contain the exact value of "Bob" in the First Name field are returned by the search (for example, the individual Bob Smith).

If you enter Bob\* as the search string, then those individuals that contain the value "Bob" followed by any other characters in the First Name field returned by the search, (for example, the individual Bobbi Smith).

If you enter \*Bob as the search string, then those individuals that contain the value "Bob" preceded by any other characters in the First Name field are returned by the search, (for example, the individual Jim-Bob Walton).

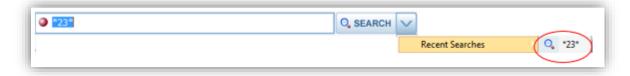
A contextual search does not distinguish between identical values in different columns. For example, if an individual smart list also displays the Last Name Column, and you enter Bob\* as the search string, then not only is the individual Bobbi Smith returned, but also, the individual James Bobby, where Bobby is contained in the Last Name field. A contextual search is available on every window (Individuals, Pedigrees, Samples, Fields, Containers, and so on) that is opened from the navigation bar on the Progeny main window.

Figure 8-8: Example of an Individuals contextual search field



The application "remembers" the last search string entered for a contextual search. To view and/or select this string, click the drop-down arrow next to the Search icon.

Figure 8-9: Selecting last contextual search string



### To carry out a contextual search

- 1. In the appropriate contextual search field, do one of the following:
  - a. Enter new search criteria or select the drop-down arrow next to the Search icon to select the last search string entered.
  - b. Click on the drop-down arrow next to the Search icon and select the last search string.



- 2. Click the Search icon. Search.
- 3. The smart list is updated with only those items that meet the search criteria.

### Main Window Queries

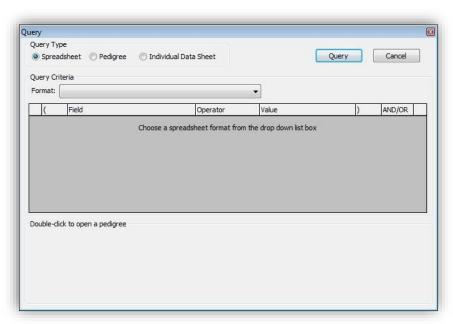
The Main Window Query function allows you to retrieve all queries that were saved as part of a spreadsheet format, run these queries, and display the query results according to the selected query type.

- Spreadsheet The results of the query are displayed in a spreadsheet format.
- Pedigree A list of pedigrees that meet the query criteria are displayed. You can double-click a pedigree to open it.
- Individual Datasheet A list of individual datasheets for all individuals who meet the query criteria is displayed. You can double-click an individual datasheet to open it.

#### To carry out the Main Window query

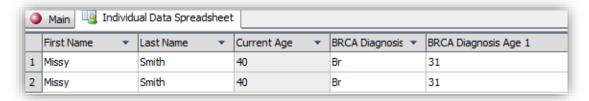
- 1. On the Progeny main window navigation bar, click the Individuals button or the Pedigrees button. The Individuals window or Pedigrees window opens, respectively.
- 2. On the window toolbar, click the Query button Query. The Query window opens.

Figure 8-10: Query window



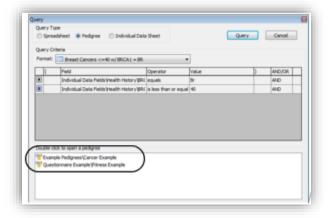
- 3. Select the Query Type.
- 4. On the Query Criteria dropdown list, select the query that is to be run.
- 5. The Query window is updated with the query criteria (Field, Operator, and so on). Click Query.
- 6. The query is run, and the results are according to the selected query type.
  - a. Spreadsheet The results of the query are displayed in a spreadsheet format.

Figure 8-11: Main window query, Spreadsheet query type



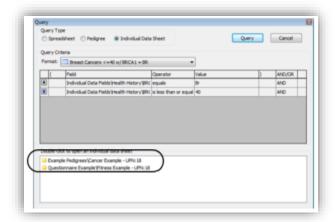
b. Pedigree – A list of pedigrees that meet the query criteria are displayed in the bottom pane of the Query window. You can double-click a pedigree to open it.

Figure 8-12: Main window query, Pedigree query type



c. Individual Datasheet – A list of individual datasheets for everyone that meets the query criteria is displayed in the bottom pane of the Query window. You can double-click an individual datasheet to open it.

Figure 8-13: Main window query, Individual Datasheet query type



### Chapter 9 Database Auditing

Database auditing involves observing a database so that you can be aware of the actions of the database users. Database auditing is often for security purposes, for example, to ensure that information is not accessed by those without the permission to access it, or that only acceptable and allowed changes were made to the data. You use the auditing feature in Progeny to track changes that were made to data in a Progeny database. You can turn the auditing feature on for specific pedigree data fields, individual data fields, and sample fields. After the audit feature is turned on, any changes that are made to the data in these fields for a pedigree, individual, or sample is tracked in the Audit report. The Audit report displays item type, the item name, the old field value, the new field value, the username for the user who changed the value and a transaction date and time stamp.

This chapter covers the following topics:

- Activating the Auditing Feature
- Generating an Audit Report



All functions that are detailed in this chapter are available only to the Progeny administrator.

### **Activating the Auditing Feature**

You use the auditing feature in Progeny to track changes that were made to data in a Progeny database. Activating the audit feature is a two-step process. You first must set the audit status for specific data fields. You can set the audit status for pedigree data fields, individual data fields, and sample fields. After you set the audit status for the data fields, you must turn on the auditing feature for the pedigrees, individuals, or samples that use these fields.



This section details the activation of the auditing feature for samples, but by analogy, you can use this procedure to activate the auditing feature for individuals and pedigrees.

### To set the audit status for data fields

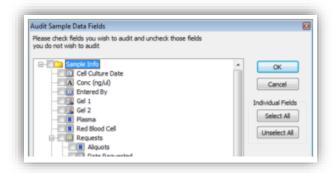
1. On the Progeny main window navigation bar, click the Fields button. • The Fields window opens.

Figure 9-1: Fields window



- 2. In the left pane of the window, open the individual database folder that contains the fields for which you are setting the audit status.
- 3. In the right pane of the Fields window, select the field or fields (CTRL-click to select multiple fields) for which you are setting the audit status.
- 4. Right-click on the selected fields, and on the context menu that opens, click Update Audit Status. The Audit Data Fields dialog box opens.

Figure 9-2: Audit Data Fields dialog box



5. Select the checkbox next to each field that is to be audited. Conversely, clear the checkbox next to each field that is not to be audited.



To select all fields for auditing in a single step, click Select All. Conversely, to remove all fields from auditing, click Unselect All.

- 6. Click OK. A message opens indicating the number of fields that were updated successfully.
- 7. Click OK to close both the message and the Audit Data Fields dialog box and return to the Fields window.
- 8. Continue to "To turn on auditing for an individual, pedigree, or sample" below.

### To turn on auditing for an individual, pedigree, or sample

- 1. On the Progeny main window navigation bar, click the Samples button it to open the Samples window.
- 2. In the left pane of the window, open the data folder that contains the samples that are to be audited.
- 3. In the right pane of the folder, select the sample or samples (CTRL-click to select multiple samples) that are to be audited.
- 4. Right-click on the selected samples, and on the context menu that opens, click Audit Status and then click On. A message opens indicating that the audit status was successfully changed.
- 5. Click OK to close the message and return to the Samples window.
- 6. Any changes made to the selected data fields for the selected samples are recorded in the Audit report.

### Generating an Audit Report

After the auditing feature is turned on, any changes that are made to the data in selected fields for a pedigree, individual, or sample are tracked in the Audit report. The Audit report displays the following data in a spreadsheet format—the item type, the item name, the old field value, the new field value, the username for the user who changed the value and a transaction date and time stamp.

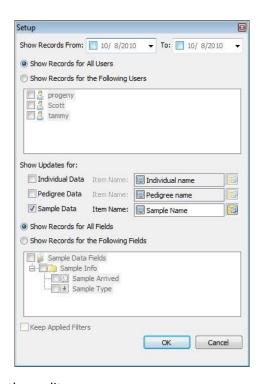
#### To access the Audit table



The following procedure details how to access the Audit table for samples, but by analogy, you can apply this procedure to individuals and pedigrees.

- 1. On the Progeny main window navigation bar, click the Samples button to open the Samples window.
- 2. On the main menu, click Administration > Field Audit Report. The Audit Setup dialog box opens.

Figure 9-3: Audit Setup dialog box

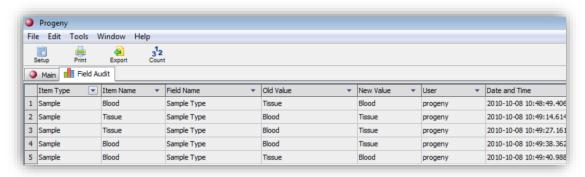


1. Specify the information for the audit.

Option	Description
Date	The ending date can be the same as the beginning date, or you can specify a date range.
Show Records for All Users     Show Records for the Following Users	If you select Show Records for the Following Users, you can select more than one user.
Show Updates:	By default, the data type that is selected is based on the window from which you opened the Audit Setup dialog box. For example, if you opened the dialog box from the Samples window, then Sample Data is selected.
	Indicate how you want to identify/refer to the item for which you are generating the Audit report. This information is displayed in the Item Name column (second column) of the Audit report.
	Individual Data - Individual Name (The default value, which is the recommended value).
	<ul> <li>Pedigree Data—Pedigree Name (The default value, which is the recommended value).</li> <li>Sample Data—Sample Name or Barcode Number.</li> </ul>
Show Records for All Fields     Show Records for the Following Fields	The fields that are available are based on the selected data type.
	Select this option to display all fields for which auditing is turned on in the Audit report.
	Select this option, and then select the specific fields for which auditing is turned that are to be displayed in the Audit report.
Keep Applied Filters	Enabled only after you generate the initial report. After you generate the initial report, select Keep Applied Filters, filter one or more columns of data, and then click Run again to generate the report with the applied filters.
	Note: Make sure to clear this option if you do not want to generate the report with the applied filters.

2. Click OK. The Audit table is generated in a spreadsheet format.

Figure 9-4: Sample Audit table



- 3. Optionally, after you generate an audit report, you can do one or more of the following:
  - a. Sort and/or filter the audit data. See Sorting and Filtering Spreadsheet Data.
  - b. Click the Setup button Setup to open the Audit setup dialog box, modify the report settings, and generate the report again.
  - c. Click the Print button to print the Audit report.
  - d. Click the Export button to export the Audit report. See Exporting Spreadsheet Data.
  - e. Click the Count Button to count the number of individuals, pedigrees, or samples (that is, the number of rows) in the spreadsheet. See Counting Spreadsheet Data.

## Chapter 10 – Backing Up and Restoring Progeny Databases

Backing up your Progeny databases is perhaps one of the most critically important things you need to do. Depending on how you use Progeny (server-enabled vs. standalone) there are several methods for accomplishing this. The following documentation will guide you through the process of setting up a bullet-proof backup system to ensure the safety of your data and show you how to restore your databases from these backups should a disaster occur.

This chapter covers the following major topics:

- Backing Up a Network (Server-Enabled) Database
- Backing Up a Standalone (Local) Database

## Backing Up a Network (Server-Enabled) Database

The Progeny Server Configuration Utility allows you to set "events" which will automatically create a copy of the database and transaction log at the time the event runs. You can have as many events as you like, but each event MUST have its own folder assigned – instead of creating dozens of copies of the database and transaction log files, the events will overwrite the previous copy each time they run to minimize disk space.

You can create these events either through the Progeny Client application or directly through the Progeny Server Configuration Utility. We will cover both methods – however, as the method for restoring a database from these backups is identical the restoration instructions will come at the end of this section.



NOTE: The Network Backup methods should only be performed by someone with access to both the server desktop and the 'progeny' superuser ID.

### Network Backups from the Progeny Client

The first step in setting up a backup system for your network databases is to create the appropriate folder structure. You will need to create a backup folder for each database, and inside each folder, you will need a sub-folder for each backup event. On Progeny's internal servers, we have a folder in the root C:\ folder called ProgenyDatabases where we keep all active and backup databases. Within this ProgenyDatabases folder, we have two sub-folders: ActiveDatabases and DatabaseBackups. All operational databases are kept within the ActiveDatabases folder and the DatabaseBackups folder contains a sub-folder for each database in the ActiveDatabases folder. Finally, within each database sub-folder of DatabaseBackups is a folder for each of the three backup events we configure. This structure is depicted below:

Figure 10-1: Automated Backup Folder Structure Workflow



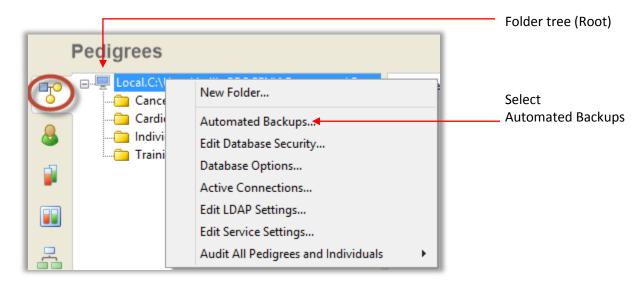


NOTE: As you are creating your folder structure, take note of the folder paths for each backup event folder for each database. You will need this when configuring your backup events.

It is recommended when creating the backup folders that you create at least two events for each database: one daily backup and one sequential backup that runs every couple of hours. When naming your backup folders, you should choose a name that starts with a letter and does not contain any spaces.

Once your folder structure is in place, launch the Progeny client either from your desktop or from the server (whichever is installed) and log into the network database using the 'progeny' superuser ID. Right-click the folder tree root (look for the little monitor icon) and select Automated Backups...

Figure 10-2: Automated Backups option within folder root menu



The Automated Backups menu manages all backup events for the network database. To create a new event, click "New" and you will be prompted to create a name for your new backup event. This name should correspond to one of the backup event folders you created for this database on the server.

Figure 10-3: The Automated Backups Menu

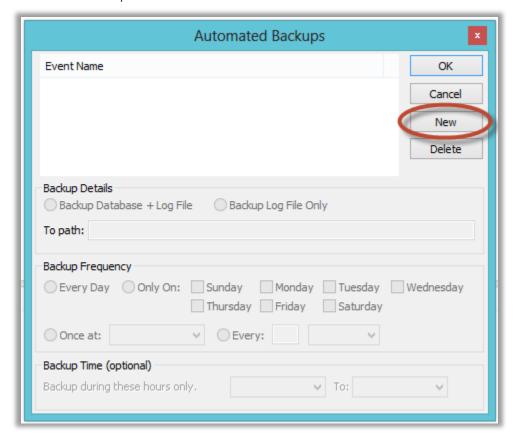


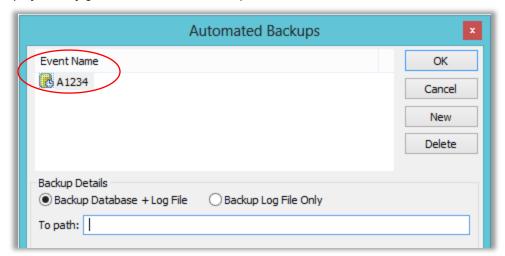
Figure 10-5: Creating A New Automated Backup Event



Once you have a name in place, you will choose the backup type and enter the folder path for the backup. You have two options here: "Backup Database + Log File" and "Backup Log File Only."

(When naming your backup folders, you should choose a name that starts with a letter and does not contain any spaces. The example above shows " $\underline{A}$ " 1234)

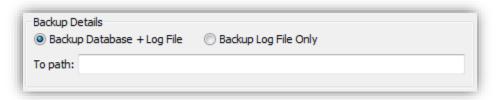
Figure 10-6: Display of a Configured Automated Backup Event





Unless you specifically need a backup that contains only the log file, make sure to choose the "Backup Database + Log File" option.

Figure 10-7: Configuring Automated Backup Type and Location

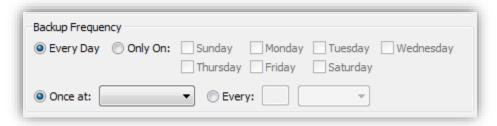


The exact folder path, *relative to the server*, will be inserted in the "To Path:" field. If this was a daily backup on one of Progeny's internal servers, we would configure this to be:

C:\ProgenyDatabases\DatabaseBackups\Progeny9Demo\Daily

Next, you will choose the backup frequency. There are several options here, so we'll cover them individually.

Figure 10-8: Automated Backup Frequency



- Every Day: Database event will run seven (7) days a week.
- Only On: Database event will only run on the days specified (checked)
- Once At: The backup event will run once per day, at the chosen time
- Every: The backup event will run repeatedly during each day, according to the number of minutes/hours specified

Finally, if you choose to have the database run more than once per day (using the **Every**: option), you can set limiting hours on the backup event using the "Backup Time (Optional)" section. The backup event will begin running at the first specified hour and will stop running at the second specified hour.

Figure 10-9: Limiting Automated Backups to Specific Hours



When you have completed your configuration of the backup event, click the OK button to commit the event. You should immediately re-open the Automated Backups menu to verify that the backup event was successfully created. If the event shows in the Event Name field (like below) then you know the backup event was successfully created.

Figure 10-10: A Configured Backup Event Shown in the Event List



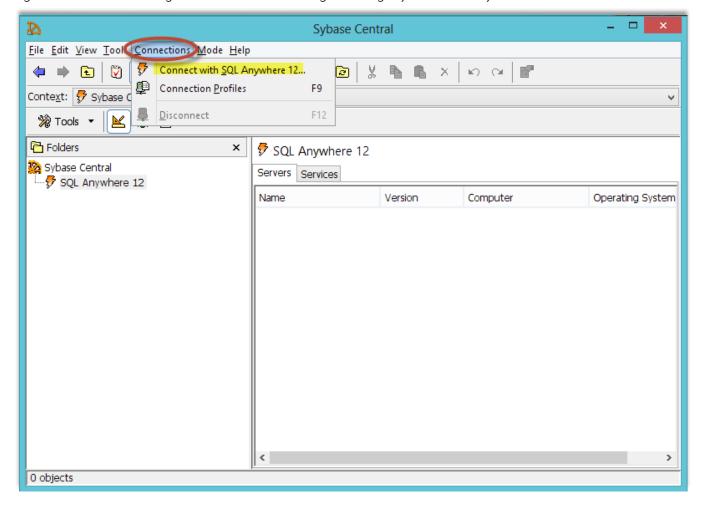
### Network Backups from the Progeny Server Configuration Utility

The 'progeny' superuser can create database backup events directly from within the Progeny Server Configuration Utility by logging directly into the database backend. Open the Progeny 9 Server Configuration Utility and choose Connections → Connect with SQL Anywhere 12... from the top menu.

Figure 10-11: Progeny Server Application Desktop Icon

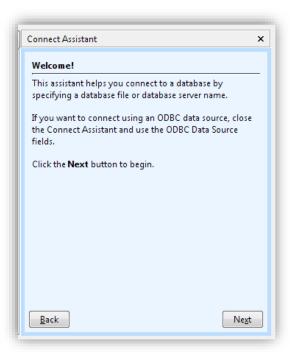


Figure 10-12: Connecting to Your Database Using the Progeny Server Utility



Once in the Connect menu, you can enter the connection information using the information tabs if you like, but the simplest method for making a database connection is to use the Connect Assistant (the light blue area on the right side of the Connect menu). Follow the prompts that the Connect Assistant provides, and when asked for a User ID and Password you will need to use the 'progeny' superuser ID and password.

Figure 10-13: Using the Connect Assistant



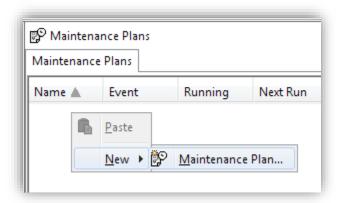
When you are successfully connected to the database backend, you will be shown a screen that contains all the administrative-level functions and options within the database. Using the Folders pane on the left-hand side scroll to the bottom of the list and select Maintenance Plans from the list to view any active Maintenance Plans (Backup Events). If you have previously configured a backup event from within the Progeny Client, you will see it listed on the right-hand side; otherwise, the area on the right will be empty.

Figure 10-14: Selecting the Maintenance Plans Option



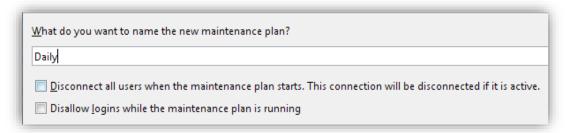
On the right-hand side, right-click the empty space and choose New → Maintenance Plan...

Figure 10-15: Creating a New Maintenance Plan



Enter a name for your Maintenance Plan (Backup Event). Unless your database file is very large (>2GB), there is no need to choose either of the two options shown below the Maintenance Plan name prompt.

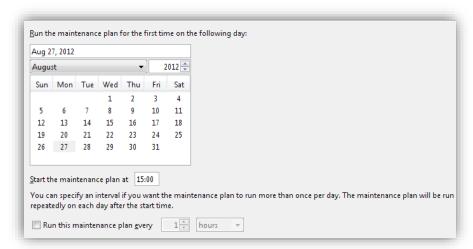
Figure 10-16: Naming Your Maintenance Plan



On the next screen, you will choose the start date and initial running time for the Maintenance Plan (Backup Event).

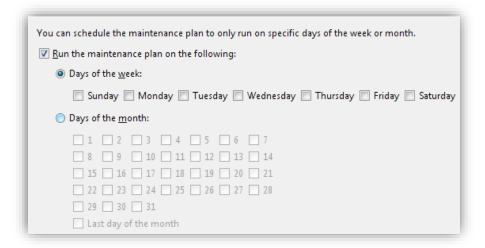
- If this Maintenance Plan will be running only once per day, the time you set here is the time when the daily backup will run.
- If this Maintenance plan will run on a repeating (sequential) basis, choose the option at the bottom to specify how often the backup will occur. The Maintenance Plan will first run at the time specified then continue according to the scheduled timeframe you set.

Figure 10-17: Selecting the Starting Date for Your Maintenance Plan



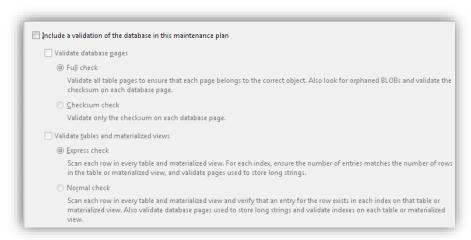
Next, you will choose which days of the week the Maintenance plan will run. Here you can select specific days of the week, have it run seven (7) days a week (by choosing all days Sunday-Saturday) or select specific days of the month for it to run.

Figure 10-18: Configuring the Frequency of Your Maintenance Plan



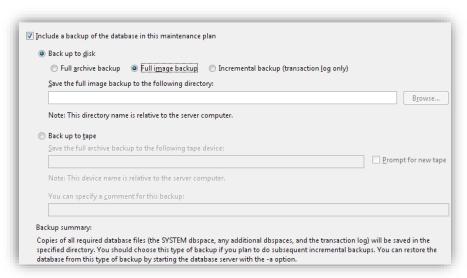
On the following page, you have the choice to include a database validation when running the Maintenance Plan. This is entirely optional, but Progeny does not recommend that you bother with this page. Running validations with your Maintenance Plan will not gain you much in terms of database safety and will make the Backup Events take significantly more time to complete.

Figure 10-19: Validating Your Maintenance Plan



After the Database Validation page, you will choose the type of backup and the backup location. Here you can choose to back up the database to the local disk or directly to a tape drive (if you have one). When choosing the type of disk backup, you should always choose "Full image backup" unless you specifically need another type. The "Full image backup" is the complete backup option. Next, you will choose a location for the backup file. Remember, this file path is relative to the server, not the desktop.

Figure 10-20: Selecting the Maintenance Plan Backup Type and Location



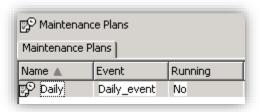
On the last page, you will have the option to have Maintenance Plan reports created and sent automatically per event. This is entirely optional and can be a royal pain depending on your network & security configurations so use this only if necessary.

Figure 10-21: Configuring Maintenance Plan Reporting



This is the final page of the Maintenance Plan configuration, so click "Finish" when complete. When the Create Maintenance Plan Wizard closes, you will see your new backup event listed on the right-hand side of the window.

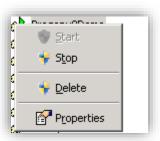
Figure 10-22: A Configured Maintenance Plan



#### Restoring a Progeny Database from a Network Backup

Should your server crash or the database become corrupted or otherwise unusable, the first step is to stop the database service. Open the Progeny Server Configuration Utility, click SQL Anywhere 11  $\rightarrow$  Services Tab, then right-click the database service in question and choose Stop.

Figure 10-23: Stopping a Progeny Database Service



Once stopped, you will see the icon next to the database service showing a red square.

Figure 10-24: A Stopped Progeny Database Service

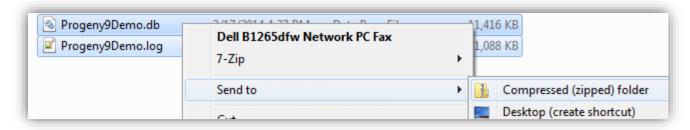




Don't close this window yet – you will be coming back to this in a few steps to restart the database service!

Navigate to the backup event folder you want to restore to and select both the database and log files. Right-click the selected files then choose, Send to  $\rightarrow$  Compressed (Zipped) Folder.

Figure 10-25: Compressing A Backup Database for Restoration



Once the Zip file has been created, copy it. Go to the folder containing your active databases and either delete the database & log files or move them to another folder for later analysis. Paste the Zip file into this folder, then right-click the Zip file and choose Extract All...



NOTE: Often the Extract All process will want to create a new folder for the extracted files using the Zip file name as the name of the new folder. In the current example Extract All would want to extract the files to the following folder:

C:\Users\%USERNAME%\Documents\Progeny 9\Progeny9Demo

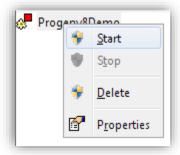
If you remove the last part of this file path (\Progeny9Demo) the database and the log file will be placed in the original folder they came from.

Now you cannot immediately restart the network database service – these backup files need to be activated first.

- If you have the Progeny Client installed on the server, launch the application and *locally* log into the database. To do this, make sure the Local Database radio button is chosen, then click Browse and navigate to the database file. Log in using the 'progeny' superuser ID and password. Once you get to the Main screen, you can close the application as the database is now active.
- If you do not have the Progeny Client installed on the server, you will need to copy the Zip file down to a desktop that has the Client application installed. Extract the database and log files, then use the instructions in the bullet point above to activate the database. Once active, put the newly-activated database and log files into a NEW Zip file, then copy this new Zip file back up to the server and Extract the activated files into the folder where you keep your active databases.

Once the activated database and log files are in place, go back to the Progeny Server Configuration Utility, right-click the database service and click Start. When the icon next to the database service name turns green, your database is active on the network and can be logged into once again.

Figure 10-26: Starting a Progeny Database Service



# Backing Up a Standalone (Local) Database

Due to HIPAA regulations regarding the storage of PHI data at the desktop level, the standalone version of Progeny is no longer available. If you are an existing user with a standalone copy of Progeny, please refer to the link below for instructions on backing up your database:

http://www.progenygenetics.com/documentation/userguides/9/Progeny9StandaloneBackups.pdf

# Section 2 – Progeny Clinical

This section will cover the creation and manipulation of pedigrees, individuals, and samples within the Clinical module. It will also explain how to customize your pedigrees and set up template features within your data folders.

This section contains the following chapters:

- 1. Progeny Clinical Terms and Conventions
- 2. Managing Data Folders
- 3. Managing Individuals
- 4. Managing Pedigrees
- 5. Drawing and Customizing Pedigrees
- 6. Managing Samples

# Chapter 1 – Progeny Clinical Terms and Conventions

Progeny Clinical is pedigree and clinical data management software that is ideal for family-based studies. The application provides the ability to draw and manage pedigrees and associated data and the ability to manage and track individuals and their associated data, whether the individual is a member of a pedigree or is a singlet (not a member of a pedigree). Progeny Clinical uses conventions in both its terminology and its output to ensure consistency and accuracy when you are manipulating and analyzing clinical data.

This chapter covers the following topics:

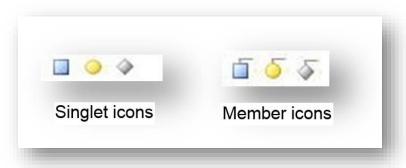
1. Progeny Clinical Terms and Conventions

### **Progeny Clinical Terms and Conventions**

Progeny Clinical provides the ability to draw and manage pedigrees and associated data and the ability to manage and track individuals and their associated data. Progeny Clinical uses conventions in both its terminology and its output to ensure consistency and accuracy when you are manipulating and analyzing clinical data.

• Individual — An individual is a single, distinct entity for which data is collected according to an approved protocol. The data can be obtained in a variety of methods, including, but not limited to physically manipulating the individual, taking samples from the individual, interviewing the individual, and so on. An individual can be a singlet, which means that the individual is not a member of a pedigree, or the individual can be a member of a pedigree. A square icon indicates a male individual, a circle icon indicates a female individual, and a diamond icon indicates an individual of unknown/undetermined gender. Variations of these icons differentiate between singlets and individuals who are a member of a pedigree.

Figure 1-1: Individual icons

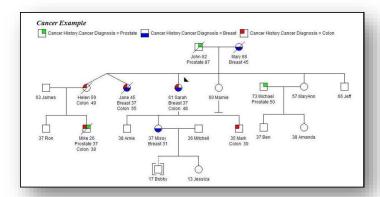


Pedigree—A pedigree, also known as a genogram, is a diagram that depicts individuals who are related by blood or another factor. Different icons represent different types of individuals. For example, a square icon indicates a male individual, a circle icon indicates a female individual, and a diamond icon indicates an individual of unknown/undetermined gender. Symbols are applied to icons to graphically represent data about the individual, such as individuals with a cancer diagnosis that equals Breast. A legend identifies each symbol. The lines connecting individuals identify the relationship between two individuals, such as marital status. The icon subtext, which is the text that is displayed above or below an individual icon on a pedigree,

is specific for the individual. The following icon indicates a pedigree:

212

Figure 1-2: Example of a Cancer History pedigree



Sample—A sample is a part that is representative of a whole or a set of elements that are drawn from and analyzed to estimate the characteristics of a population. A sample can be an unassociated sample, or it can be associated with an individual. The following icon indicates a sample: .

### Chapter 2 – Managing Data Folders

Data folders are the folders that house the data for individuals, pedigrees, samples, and containers. One of the key features of Progeny Clinical is the use of template data folders to organize individuals, pedigrees, and samples. When you create a new individual or pedigree, you must set specific options for the individual or pedigree—the datasheet layout, the pedigree datasheet layout, the icon subtext, and so on, and you must assign the individual or a pedigree to a data folder. Similarly, when you create a new sample, you must specify the sample datasheet layout. If the data folder is a template data folder, then these options are set at the folder level. Every new individual, pedigree, or sample that you assign to the data folder is assigned with the same template folder options, which prevents you from having to assign the options each time that you create an individual, pedigree, or sample. Because the first step in creating a new individual, pedigree, or sample is the selection of its data folder, the creation, and management of data folders must be discussed even before the creation of a new individual, pedigree, or sample.

#### This chapter covers the following topics:

- Adding Individual and Pedigree Data Folders
- **Adding Sample Data Folders**
- Copying Template Options Between Data Folders or Items
- Modifying and Deleting Data Folders

### Adding Individual and Pedigree Data Folders

When you add a new individual or pedigree data folder, you have the option of adding the folder as a template folder or as a non-template folder. A template data folder replicates the same properties (the datasheet format, the pedigree datasheet format, the icon subtext, and so on) for all the individuals or pedigrees that are contained in the data folder. Every new individual or pedigree that you add to the data folder is added with the same template folder options, which prevents you from having to assign the options each time that you create an individual or pedigree. If you add a non-template data folder, which is a folder without a single template option selected, then you must assign these options each time that you add an individual or pedigree to the data folder.

When you add a new individual data folder, an identically named pedigree data folder with the same template options is added. Likewise, when you add a new pedigree data folder, an identically named individual data folder with the same template options is added. This action allows you to include singlets with individuals who are members of a pedigree in the same folder and it also allows you to view and work with individuals who are members of a pedigree separately from the pedigree. In addition, individual data folders and pedigree data folders are linked. Any changes that you make to the options for a data folder on the Individuals window are also made to the same folder on the Pedigrees window and vice versa.



Progeny Software highly recommends that you create template folders whenever possible; however, in the rare event that you want to use the Copy feature to carry out "one-off" copies of individuals, pedigrees, or samples in a data folder, you can create a non-template folder. See <a href="Copying Template Options Between Data">Copying Template Options Between Data</a> Folders or Items.

### To add an individual or pedigree data folder



The following procedure details the creation of an individual folder, however, by analogy, you can follow this procedure to create a pedigree folder.

1. On the Progeny main window navigation bar, click the Individuals button window.

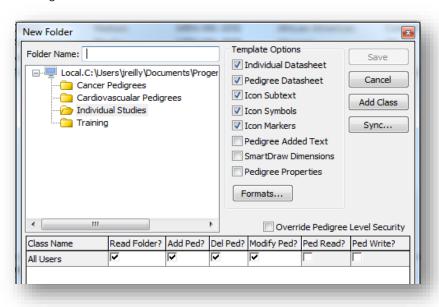


to open the Individuals

2. Select the location for the new folder – this can be stored at the root level (which is the database that you are currently logged in to), or it can be a sub-folder of another folder. Should you create a folder in the wrong location or change your mind about the layout you can simply drag a folder to move it to a different location.

- 3. On the Individuals window toolbar, click the New Folder button. New Folder. The New Folder dialog box opens.
- 4. The dialog box contains a list of options that you can set for a template folder. In addition, if you are the Progeny administrator, the dialog box contains options for setting folder security.

Figure 2-1: New Folder dialog box



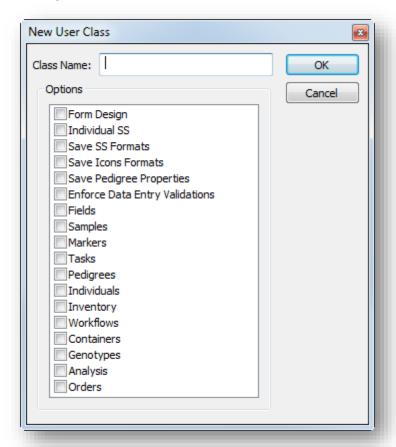
- 5. In the Folder Name field, enter a name for the new data folder.
- 6. Do one of the following:
  - a. If you are creating a non-template folder, then clear all the template options on the New Folder dialog box.
  - b. If you are creating a template folder, then specify the pertinent template options for the data folder.

Option	Description
Individual datasheet	Selected by default. The same individual datasheet format is used for entering and storing individual data for the individuals who are in the folder.
Pedigree datasheet	Selected by default. The same pedigree datasheet format is used for entering and storing pedigree data for the pedigrees that are in the folder.
Icon Subtext	Selected by default. The same icon subtext, which is the text that is displayed above or below an icon on a pedigree, is displayed for every icon on all pedigrees that are in the folder. See <a href="Configuring Pedigree">Configuring Pedigree</a> <a href="Loos">Loos</a>
Icon Symbols	Selected by default. The same icon symbols, which are symbols that graphically represent data about on an individual on a pedigree, are used for all pedigrees in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a>
Icon Markers	Selected by default. The same icon markers, which are the color codes that are used to identify genotypes (marker values) in pedigrees, are used for all pedigrees in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a> Note: This feature is a Progeny Lab feature and requires the Marker module to be used in Progeny Clinical.
Pedigree Added Text	The same text (header, footer, and/or date) is displayed on the pedigrees that are in the folder. See <a href="Pedigree Properties">Pedigree Properties</a>
SmartDraw Dimensions	All the pedigrees that are in the folder use the same drawing dimensions such as spacing, icon size, and so on. See Pedigree Properties
Pedigree Properties	All options from the Progeny and Pedigree tabs of the Properties menu are shared among the pedigrees in this folder. See Pedigree Properties

7. If you are the Progeny administrator, then do one or more of the following to set the security for the data folder; otherwise, continue.

- 8. Modify the security settings for All Users or for the different classes of users.
  - a. Read Folder? Open and view the contents of the data folder.
  - b. Add Ped? Add pedigrees to the data folder.
  - c. Del Ped? Delete pedigrees from the data folder.
  - d. Modify Ped? Modify pedigrees in the data folder.
  - e. Ped Read? View the data in the pedigree.
  - f. Ped Write? Write data to the pedigree.
  - g. Click Add Class to open the New User Class dialog box to create a new user class, and then set the folder security for the new class.

Figure 2-2: New User Class dialog box



9. If you have assigned security to one more pedigree in the folder, and you want to override the security settings for these pedigrees based on the security settings at the folder level, click Override Pedigree Level Security, and then set the appropriate pedigree level security for the different classes of users.

Figure 2-3: Override Pedigree Level Security Option

▼ Override Pedigree Level Security

a. Click on Formats...Button

Figure 2-4: Template Formats dialog box

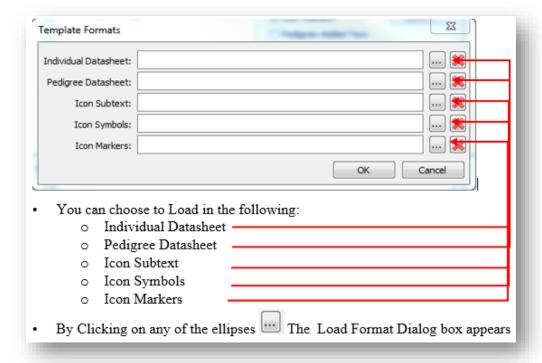
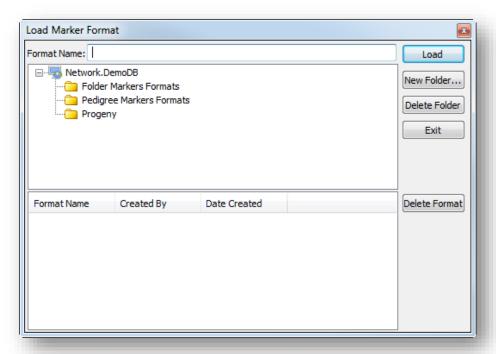
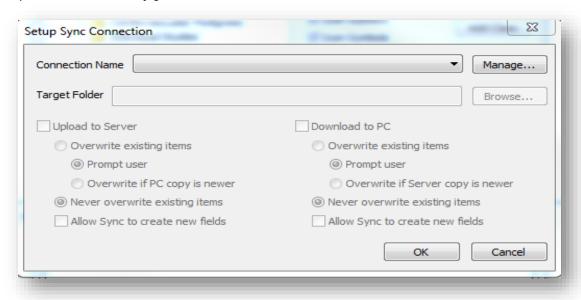


Figure 2-5: Load Marker Format dialog box



- i. It is highly recommended that you enable formats from this menu. By setting them in this place, you are putting an ironclad default in place for Progeny. ALL pedigrees and individuals within the folder will open with the formats listed in this menu as the configured defaults. A user may change this using one of the options within the pedigree viewer or individual/pedigree datasheet but every time a pedigree or individual is opened these formats will load by default.
- 10. You can choose to Sync... This allows the user to Setup a Sync Connection with Database. Click on Manage...to choose the database you want to connect to.

Figure 2-6: Sync Connection Configuration Window



- 11. Click Save. A message opens indicating the settings for the folder were successfully changed.
- 12. Click OK to close the message and return to the Individuals window.

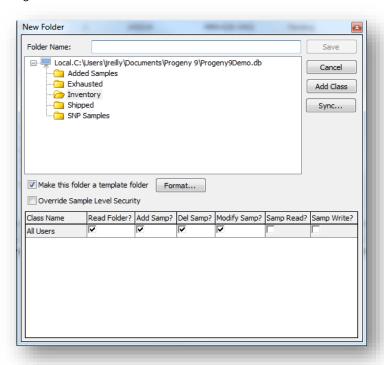
# Adding Sample Data Folders

When you add a new sample data folder, you have the option of making the folder a template folder. If you make the folder a template folder, then all samples that are added to the folder use the same sample datasheet for entering and storing data. If you do not make the folder a template folder, then each time you add a sample to the folder, you must create a new datasheet for the sample.

### To add a sample data folder

- 1. On the Progeny main window navigation bar, click the Samples button to open the Samples window.
- 2. On the Samples window toolbar, click the New Folder button. New Folder.
  - a. The New Folder dialog box opens. The dialog box contains an option for making a sample folder a template folder. In addition, if you are the Progeny administrator, the dialog box contains options for setting folder security.

Figure 2-7: New Folder dialog box



- 3. In the New Folder Name field, enter a name for the new data folder.
- 4. Optionally, to make this folder a template folder, select Make this folder a template folder.
  - a. Optionally, to load in a Sample Datasheet, select the Formats button.



If you make the folder a template folder, then all samples that are added to the folder use the same sample datasheet for entering and storing data; otherwise, each time you add a sample to the folder, you must create a new datasheet for the sample.

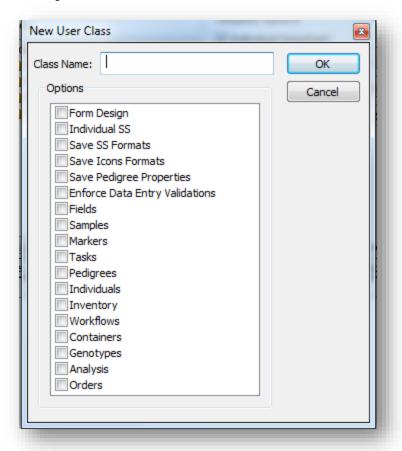
5. Select the location for the new folder. The folder can be stored at the root level (which is the database that you are currently logged in to), or it can be a sub-folder of another folder.



If needed, you can simply drag a folder to move it to a different location.

- 6. If you are the Progeny administrator, then do one or more of the following to set the security for the data folder; otherwise, continue.
- 7. Modify the security settings for All Users or for the different classes of users.
  - a. Read Folder? —Open and view the contents of the data folder.
  - b. Add Ped? —Add samples to the data folder.
  - c. Del Ped? —Delete samples from the data folder.
  - d. Modify Ped? —Modify samples in the data folder.
- 8. Click Add Class to open the New User Class dialog box to create a new user class, and then set the folder security for the new class.

Figure 2-8: New User Class dialog box



- 9. If you have assigned security to one more samples in the folder, and you want to override the security settings for these samples based on the security settings at the folder level, click Override Sample Level Security, and then set the appropriate sample level security for the different classes of users.
  - a. Ped Read? —View the sample data.
  - b. Ped Write? —Write data to the sample.
- 10. Click Save. A message opens indicating the settings for the folder were successfully changed. Click OK to close the message and return to the Samples window.

## Copying Template Options Between Data Folders or Items

You typically use the Copy feature to apply the same template options to all items (individuals, pedigrees, or samples) that are in the same data folder. The source item is the item (individual, pedigree, or sample) from which you are copying the template options. The destination folder is the data folder to which you are copying the template options. The destination folder must contain at least one item (individual, pedigree, or sample) and at least one template option must be selected for the folder for the template options to be copied.



Although less common, you can also use the Copy feature to copy template options to a single individual, pedigree, or sample that is in a non-template data folder without affecting the options that are set for the other items in the folder. If you use the Copy feature to copy template options to a single individual, pedigree, or sample that is in a template data folder, then the template options are applied to all items in the folder.

To copy template options to an individual or pedigree data folder



The following procedure details how to copy template options to an individual data folder, but by analogy, you can follow this procedure to copy template options to pedigree data folder.

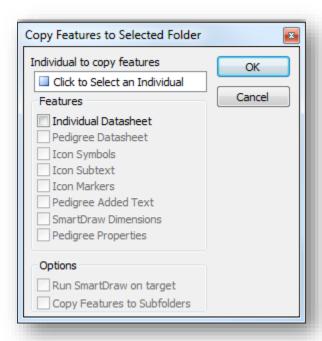


- 1. On the Progeny main window navigation bar, click the Individuals button window.
- to open the Individuals
- 2. Right-click on the destination folder (which is the individual data folder to which you are copying the template options), and on the context menu that opens, click Copy Features to Folder. The Copy Features to Selected folder dialog box opens.



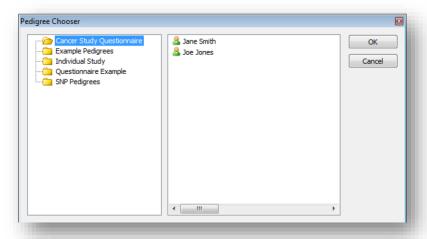
Remember, the destination folder must contain at least one item (individual or pedigree) for the template options to be copied.

Figure 2-9: Copy Features to Selected Folder dialog box



3. Click in the field labeled Click to Select an Individual. The Pedigree Chooser dialog box opens.

Figure 2-10: Pedigree Chooser Window

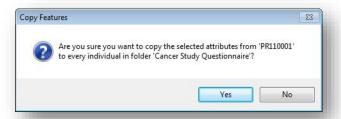


- 4. In the Pedigree Chooser dialog box, select the source individual from whom you are copying the template folder options, and then click OK. The Pedigree Chooser dialog box closes, and you are returned to the Copy Features to Selected folder dialog box.
- 5. Select the source template options that are to be copied.

Option	Description
Individual datasheet	Available for both individual data folders and pedigree data folders. The same individual datasheet format is used for entering and storing individual data for the individuals who are in the folder.
Pedigree datasheet	Available only for pedigree data folders. The same pedigree datasheet format is used for entering and storing pedigree data for the pedigrees that are in the folder.
Icon Subtext	Available only for pedigree data folders. The same icon subtext, which is the text that is displayed above or below an icon on a pedigree, is displayed for every icon on all pedigrees that are in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a>
Icon Symbols	Available only for pedigree data folders. The same icon symbols, which are symbols that graphically represent data about on an individual on a pedigree, are used for all pedigrees in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a>
Icon Markers	Available only for pedigree data folders. The same icon markers, which are the color codes that are used to identify the genotypes (marker values) in pedigrees, are used for all pedigrees in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a> Note: This feature is a Progeny Lab feature and requires the Marker module to be used in Progeny Clinical.
Pedigree Added Text	Available only for pedigree data folders. The same text (header, footer, and/or date) is displayed on the pedigrees that are in the folder. See <a href="Pedigree Properties">Pedigree Properties</a>
SmartDraw Dimensions	Available only for pedigree data folders. All the pedigrees that are in the folder use the same drawing dimensions such as spacing, icon size, and so on. See <a href="Pedigree Properties">Pedigree Properties</a>
Pedigree Properties	All options from the Progeny and Pedigree tabs of the Properties menu are shared among the pedigrees in this folder. See Pedigree Properties
Run SmartDraw on target	Available only for pedigree data folders. Select this option to run  SmartDraw on all the pedigrees in the selected destination folder.
Copy Features to SubFolders	Available for both individual and pedigree data folders. Select this option to copy the selected template options not only to the selected destination folder but also, to all its subfolders.

6. Click OK. A Copy Features message opens, asking you if you are sure that you want to copy the selected attributes from the individual that you selected in Step 5 to every individual in the destination folder that you selected.

Figure 2-11: Copy Features message



- 7. Click Yes. A Copy Progress dialog box opens, indicating the status of the copy progress.
- 8. After the copy process is completed, click Close in the Copy Progress dialog box.
- 9. The Copy Progress and Copy Features to Selected folder dialog boxes close. You return to the Individuals window. The selected template options are copied at the data folder level. All individuals that you add to the folder will now use the same datasheet format.

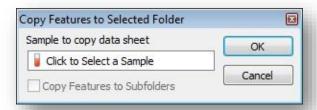
### To copy a sample datasheet format to a sample folder

- 1. On the Progeny main window navigation bar, click the Samples button to open the Samples window.
- 2. Right-click on the destination folder (which is the sample data folder to which you are copying the datasheet format), and on the context menu that opens, click Copy Features to Folder.
- 3. The Copy Features to Selected Folder dialog box opens.



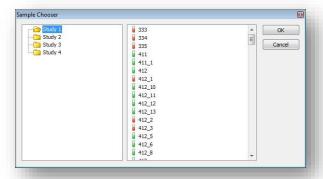
Remember, the destination folder must contain at least one sample for the datasheet format to be copied.

Figure 2-12: Copy Features to Samples dialog box



- 4. Optionally, to copy the sample datasheet format not only to the selected folder but also, to all its subfolders, select Copy Features to Subfolders.
- 5. Click in the field labeled "Click to Select a Sample." The Sample Chooser dialog box opens. This dialog box lists all the available sample data folders and the samples in them.

Figure 2-13: Sample Chooser dialog box



- 6. In the Sample Chooser dialog box, select the source sample from which you are copying the sample datasheet format, and then click OK.
- 7. The Sample Chooser dialog box closes. The name of the sample that you selected is displayed in the Copy Features to Selected Folders dialog box.
- 8. Click OK in the Copy Features to Selected Samples dialog box.
- 9. A message opens asking you if you are sure that you want to copy the datasheet format from the sample that you selected in Step 5 to the folder that you selected in Step 2.
- 10. Click Yes. The message closes. The sample datasheet format is copied at the sample folder level. All samples that you add to the folder will now use the same datasheet format.

### To copy template options to individuals or pedigrees



The following procedure describes how to copy template features to a single individual that is in a non-template folder, but by analogy, you can follow this procedure to copy template features to a single pedigree that is in a non-template folder.

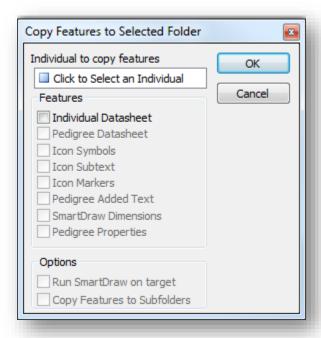
1. On the Progeny main window navigation bar, click the Individuals button window.



to open the Individuals

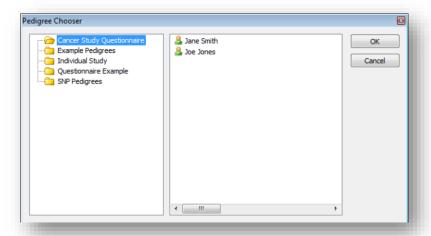
- 2. Do one of the following:
  - a. If you are copying template options to a single destination individual, (which is the individual to which you are copying the template options), then right-click on the individual and on the context menu that opens, click Copy Features to Individual(s).
  - b. If you are copying template options to multiple destination individuals, CTRL-click to select these multiple individuals, and then right-click and on the context menu that opens, click Copy Features to Individual(s).

Figure 2-14: Copy Features to Selected Individuals dialog box



3. Click in the field labeled "Click to Select an Individual." The Pedigree Chooser dialog box opens.

Figure 2-15: Pedigree Chooser

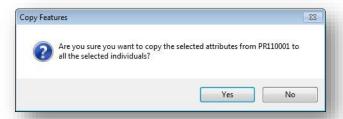


- 4. In the Pedigree Chooser dialog box, select the source individual from whom you are copying the template folder options, and then click OK.
  - a. The Pedigree Chooser dialog box closes, and you are returned to the Copy Features to Selected folder dialog box.
- 5. Select the source template options that are to be copied. A description of these options is displayed below.

Option	Description
Individual datasheet	Available for both individual data folders and pedigree data folders. The same individual datasheet format is used for entering and storing individual data for the individuals who are in the folder.
Pedigree datasheet	Available only for pedigree data folders. The same pedigree datasheet format is used for entering and storing pedigree data for the pedigrees that are in the folder.
Icon Subtext	Available only for pedigree data folders. The same icon subtext, which is the text that is displayed above or below an icon on a pedigree, is displayed for every icon on all pedigrees that are in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a>
Icon Symbols	Available only for pedigree data folders. The same icon symbols, which are symbols that graphically represent data about on an individual on a pedigree, are used for all pedigrees in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a>
Icon Markers	Available only for pedigree data folders. The same icon markers, which are the color codes that are used to identify the genotypes (marker values) in pedigrees, are used for all pedigrees in the folder. See <a href="Configuring Pedigree Icons">Configuring Pedigree Icons</a> Note: This feature is a Progeny Lab feature and requires the Marker module to be used in Progeny Clinical.
Pedigree Added Text	Available only for pedigree data folders. The same text (header, footer, and/or date) is displayed on the pedigrees that are in the folder. See <a href="Pedigree Properties">Pedigree Properties</a>
SmartDraw Dimensions	Available only for pedigree data folders. All the pedigrees that are in the folder use the same drawing dimensions such as spacing, icon size, and so on. See <a href="Pedigree Properties">Pedigree Properties</a>
Pedigree Properties	All options from the Progeny and Pedigree tabs of the Properties menu are shared among the pedigrees in this folder. See Pedigree Properties
Run SmartDraw on target	Available only for pedigree data folders. Select this option to run  SmartDraw on all the pedigrees in the selected destination folder.
Copy Features to SubFolders	Available for both individual and pedigree data folders. Select this option to copy the selected template options not only to the selected destination folder but also, to all its subfolders.

6. Click OK. A Copy Features message opens, asking you if you are sure that you want to copy the selected attributes from the individual that you selected in Step 4 to all the individuals that you selected in Step 2.

Figure 2-16: Copy Features message



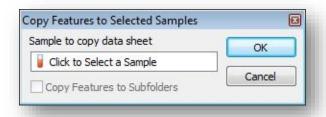
- 7. Click Yes. A Copy Progress dialog box opens, indicating the status of the copy progress.
- 8. After the copy process is completed, click Close in the Copy Progress dialog box.
- 9. The Copy Progress and Copy Features to Selected folder dialog boxes close. You return to the Individuals window. The selected template options are copied at the individual level.

#### To copy a sample datasheet format to samples



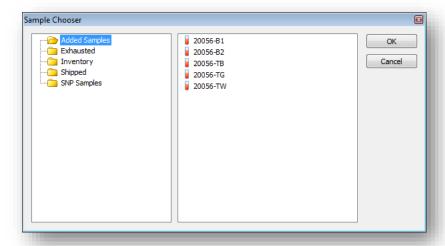
- 2. Do one of the following:
  - a. If you are copying a datasheet format to a single destination sample, (which is the sample to which you are copying the format), then right-click on the sample and on the context menu that opens, click Copy Features to Sample(s).
  - b. If you are copying a datasheet format to multiple destination samples, CTRL-click to select these multiple samples, and then right-click and on the context menu that opens, click Copy Features to Sample(s).

Figure 2-17: Copy Features to Selected Samples dialog box



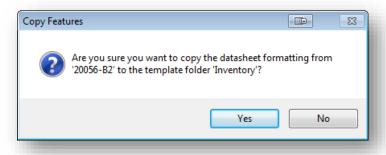
- 3. Click in the field labeled "Click to Select a Sample."
  - a. The Sample Chooser dialog box opens. This dialog box lists all the available sample data folders and the samples in them.

Figure 2-18: Sample Chooser dialog box



- 4. In the Sample Chooser dialog box, select the source sample from which you are copying the sample datasheet format, and then click OK.
- 5. The Sample Chooser dialog box closes and the name of the sample that you selected is displayed in the Copy Features to Samples dialog box.
- 6. Click OK in the Copy Features to Selected Samples dialog box.
  - a. A message opens asking you if you are sure that you want to copy the datasheet format from the sample that you selected in Step 4 to the samples that you selected in Step 2.

Figure 2-19: Copy Features confirmation dialog



7. Click Yes. The message closes. The sample datasheet format is copied at the sample level.

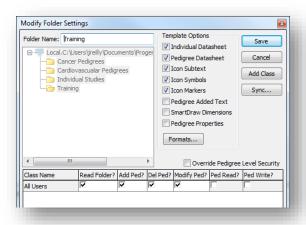
## Modifying and Deleting Data Folders

If you are the Progeny administrator, or if your administrator has set certain security levels for your user account, after you set the template options for a data folder, you can always modify the options. Likewise, after you create a non-template folder, you can always modify the folder and set template options for the folder. If you modify the template options for a data folder, then the modified options are applied only to new items (individuals, pedigrees, or samples) that you add to the folder. The modified options do not override the current template options for any existing items in the folder. In addition, if you are the Progeny administrator, you can modify the security levels for a data folder and you can delete a data folder. If you are not the Progeny administrator, then the security levels that your administrator has set for user account determine whether you can delete a data folder.

### To modify a data folder

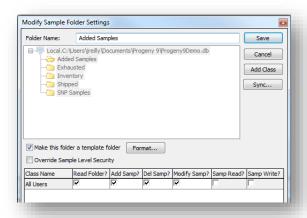
1. Right-click on the data folder that you are modifying, and on the context menu that opens, click Edit Folder. The Modify Folder Settings dialog box opens.

Figure 2-20: Modify Folder Settings dialog box for an individual or a pedigree



- 2. Modify the template options for the folder as needed.
- 3. Do one of the following:
  - a. If you are the Progeny administrator, then optionally, modify the security for the data folder as needed, and then click Save.
  - b. If you are not the Progeny administrator, then click Save. A message opens indicating the settings for the folder were successfully changed.
- 4. Click OK to close the message and return to the item (Individuals, Pedigrees, or Samples) window.

Figure 2-21: Modify Sample Folder Settings dialog box



#### To delete a data folder

When you delete a data folder, all the data that is in the folder (individuals, pedigrees, or samples) is deleted as well as any subfolders and all the data that is in the subfolders.

- 1. Right-click on the data folder that you are deleting, and on the context menu that opens, click Delete Folder.
  - a. A Confirm Delete Folder message opens that asks you to confirm the deletion of the data folder, its data, and any subfolders and all the data in the subfolders.
- 2. Click OK to confirm the deletion.
  - a. The message closes and you return to the item (Individuals, Pedigrees, or Samples) window.

# Chapter 3 Managing Individuals

An individual is a single, distinct entity for which data is collected according to an approved protocol. Managing individuals in Progeny Clinical consists of creating new individuals, creating individual shortcuts, creating datasheets for individuals, modifying an individual, creating individual shortcuts, copying, moving, and deleting individuals, moving individuals to a pedigree, and importing individual data.

This chapter covers the following topics:

- Adding a New Individual
- **Creating a New Individual Datasheet**
- Modifying an Individual
- **Creating Individual Shortcuts**
- **Copying and Moving Individuals**
- Deleting an Individual
- Moving an Individual to a Pedigree



For detailed information about importing individual data, see Appendix B, **Importing Clinical Data** 

# Adding a New Individual

When you first add an individual, the individual is not contained in a pedigree. An individual who is not contained in a pedigree is referred to as a singlet. After you create a singlet, you can move the single to an existing pedigree or to a new pedigree.

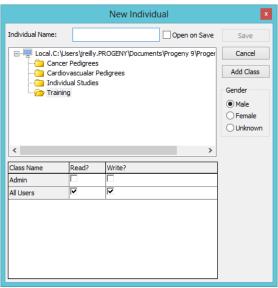
#### To add a new individual

1. On the Progeny main window navigation bar, click the Individuals button



2. On the Individuals window, click the New Individual button New Individual dialog box opens.

Figure 3-1: New Individual dialog box



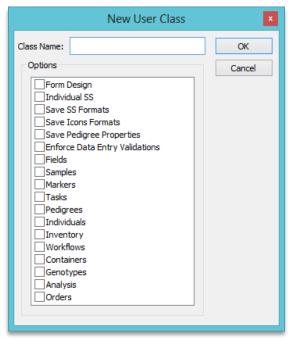
- 3. Enter the name of the new individual, select the data folder to which the individual is being added and select the gender for the individual.
  - a. Optionally, select Open on Save (once you select this, it defaults to be checked for every new individual).



The Individual Name field and Gender field are system fields. See <u>Individual System</u> Fields.

- b. If the selected folder to which you assigned the individual has an Individual Datasheet template assigned the formatted datasheet opens in Data Entry mode and you can immediately begin entering data for the individual after saving. If no datasheet format has been assigned to the folder then a blank datasheet opens in Data Entry mode. You can click Form Design mode and design the datasheet format for the folder before you add any more individuals to the folder. See <a href="Creating a New Individual Datasheet">Creating a New Individual Datasheet</a>
- 4. If you are the Progeny administrator, then do one or more of the following to set the security for the individual as described in Step 4; otherwise, continue to Step 5.
  - a. Modify the security settings for All Users or for the different classes of users.
    - i. Read? View the data (name and data in the individual datasheet) for the individual.
    - ii. Write? Write data (change the name and enter data to an individual datasheet) to the individual.
  - b. Click Add Class to open the New User Class dialog box to create a new user class, and then set the security for the new class by selecting a group of options that you will want to assign for individuals.

Figure 3-2: New User Class dialog box



- 5. Click OK. A message opens indicating the settings for the individual were successfully changed.
- 6. Click OK to close the message and return to the Individuals window.

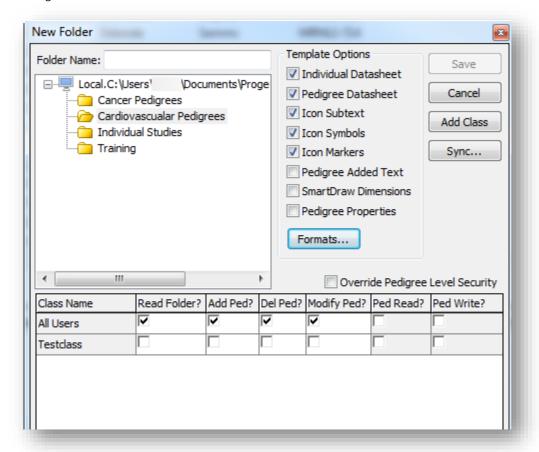
# Creating a New Individual Datasheet

You use an individual datasheet to enter and store data for an individual in a Progeny database. All the individuals who are assigned to an individual data folder can use the same datasheet format, or everyone in the folder can use a unique datasheet format. You can create and format an individual datasheet at the time that you add an individual to a data folder, or later.

#### To create an individual datasheet

- 1. Create an individual data folder.
  - a. If all the individuals who are assigned to the folder are to use the same individual datasheet format, make sure to select the Individual Datasheet template option; otherwise, clear it. See <u>Adding</u> <u>Individual and Pedigree Data Folders</u>.

Figure 3-3: Creating a new Individual Datasheet

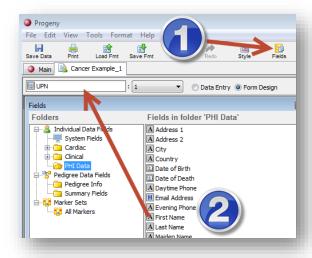


- 2. Create a new individual (see <u>Adding a New Individual</u>) within the data folder. When you are creating the individual, do one of the following:
  - a. To have a blank datasheet open immediately after you save the individual, select Open on Save. You can click Form Design on this blank datasheet and format the datasheet as needed. If the Individual Datasheet template option has been selected for the data folder, then all the individuals that you subsequently add to the folder will use the same datasheet format for entering and storing data; otherwise, you must format a datasheet for everyone that you add to the folder.
  - b. To add all individuals to the folder before you create and format the datasheet, do not select Open on Save for any individual whom you add to the folder. After you have added all the needed individuals to the folder, you can simply double-click on any individual in the folder to open a blank datasheet and format the datasheet as needed. If the Individual Datasheet template option has been selected for the data folder, then all individuals who are currently added to the folder or that you subsequently add to the folder will use this same datasheet format for entering and storing data; otherwise, you must format a datasheet for everyone in the folder.
    - i. See <u>Creating and Formatting Datasheets</u> for detailed information about creating and formatting an individual datasheet.



When you are entering data into an individual datasheet, by default, the UPN for the individual is displayed above the datasheet. To make it easier for you to identify an individual when you are entering data for the individual, you can display a different field above the datasheet, for example, the Patient ID. To change the field, in Form Design mode, click the Fields button to open the Select Field dialog box, and drag the new field onto the UPN field in the upper left corner of the individual datasheet. The new field and its data are displayed instead of the UPN number.

Figure 3-4: Changing the Individual selector within the Individual Datasheet from UPN to First Name.



# Modifying an Individual

After an individual is created, you can modify the gender of the individual. If you are the Progeny administrator, or your administrator has set Write access for the individual for your user account, you can rename the individual. As the Progeny administrator, you can also modify the security levels for the individual.

### To modify an individual

1. On the Progeny main window navigation bar, click the Individuals button window.



to open the Individuals

- 2. Open the data folder that contains the individual that is being modified.
- 3. Select the individual that is being modified, and on the main menu bar in the upper left corner (to the right of 'Files" and "Folders"), click "Individuals", and then click the appropriate option.
  - a. The options that are available from the Individuals menu depends on whether the individual is a single or a member of a pedigree.

Figure 3-5: Individuals right-click menu for a singlet

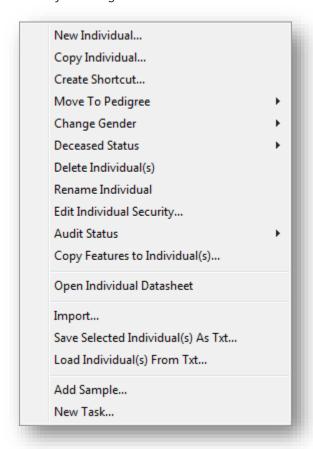
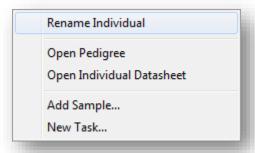


Figure 3-6: Individuals right-click menu for a pedigree member



Option	Description
Copy Individual	Available only for a singlet. Creates an exact duplicate of the individual then selects the Individual Name field for you to assign a unique name.
Move to Pedigree	Available only for a singlet. Allows the user to associate the selected singlet with an existing pedigree or to create a new pedigree for this individual.
Change Gender	Available only for a singlet. Options are Male, Female, and Unknown.  Note: You can change the gender of an individual that is included in a pedigree only from the pedigree drawing window. See Working with Individuals in a Pedigree.
Rename Individual	Automatically selects the individual name for modification.
Edit Individual Security	Available only for a singlet. Opens the Individual Security dialog box in which you can do one or both of the following:  • Modify the security settings for All Users or for specific classes.  • Read? – View the data (name and data in the individual datasheet) for the individual.
	<ul> <li>Write? – Write data (change the name and enter data to an individual datasheet) to the individual.</li> <li>Click Add Class to open the New User Class dialog box to create a new user class, and then set the individual security for the new class.</li> </ul>

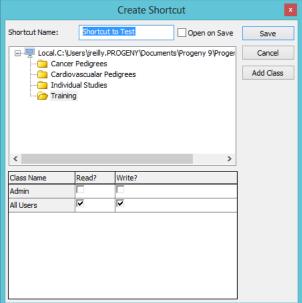
## **Creating Individual Shortcuts**

An individual shortcut is a link that points to an individual in a Progeny database. You create an individual shortcut so that you can add an individual to multiple folders without having to copy or duplicate the individual. Although a shortcut points to the same original individual, each shortcut inherits the template options of the folder in which it is contained. For example, you might place an individual shortcut in an individual data folder named Colon Cancer, which has been assigned an individual datasheet that has been specifically designed for entering data that is related to colon cancer. You can also place this same individual shortcut in an individual data folder named Breast Cancer, which has been assigned an individual datasheet that has been specifically designed for entering data related that is related to breast cancer. You can create an individual shortcut only for a singlet.

#### To create an individual shortcut

- 1. On the Progeny main window navigation bar, click the Individuals button window.
- 2. Open the data folder that contains the individual for which you are creating the shortcut.
- 3. Select the individual for which you are creating the shortcut, and on the main menu once more, click "Individuals" > Create Shortcut.
  - a. The Create Shortcut dialog box opens. The Shortcut Name field is automatically populated with a default value, but you can change this value.



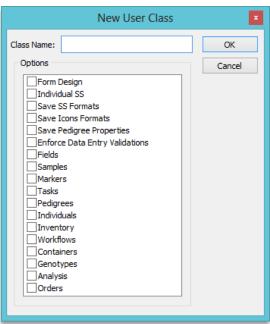


- 4. Select the folder in which to save the shortcut.
- 5. Optionally, do one or both of the following:
  - a. Change the shortcut name.
  - b. Select Open on Save.

If the Individual Datasheet template option has been selected for the folder to which you assigned the shortcut and a datasheet has been created for the folder after you save the shortcut, the formatted datasheet opens in Data Entry mode and you can immediately begin entering data for the individual. If no datasheet has been created for the shortcut, then a blank datasheet opens in Data Entry mode. You can click Form Design mode and design the datasheet format for the folder before you add any more shortcuts to the folder. See Creating a New Individual Datasheet.

- 6. If you are the Progeny administrator, then do one or more of the following to set the security for the individual; otherwise, continue to Step 7.
  - a. Modify the security settings for All Users or for the different classes of users.
    - i. Read? View the data (name and data in the individual datasheet) for the individual.
    - ii. Write? Write data (change the name and enter data to an individual datasheet) to the individual.
    - iii. Click Add Class to open the New User Class dialog box to create a new user class, and then set the individual security for the new class.

Figure 3-6: New User Class dialog box



- 7. Click OK. A message opens indicating the settings for the individual were successfully changed.
- 8. Click OK to close the message and return to the Individuals window.

# Copying and Moving Individuals

- 1. You can copy an individual between data folders, and you can move an individual between data folders. For any of these actions, the following are important points to note:
  - a. Copying an individual leaves the original individual in its original data folder while putting a copy (or duplicate) of the individual in the new destination folder. Any edits that you make to the copied individual are not reflected in the original individual.
  - b. Moving an individual removes the individual from its original data folder and places the individual in a new destination folder. A copy of the original individual is not made.

## To copy or move an individual

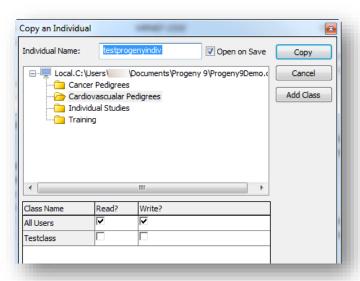


- 1. On the Progeny main window navigation bar, click the Individuals button
- 2. Open the data folder that contains the individual that you are copying or moving, and then continue to one of the following:
  - a. To copy an individual between data folders
  - b. To move an individual between data folders

#### To copy an individual between data folders

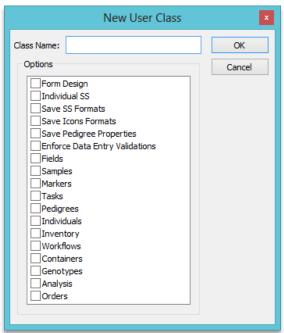
1. On the main menu, click Individuals > Copy Individual. The Copy an Individual dialog box opens.

Figure 3-7: Copy an Individual



- 2. Select the data folder to which the individual is being copied.
- 3. Optionally, before you copy the individual to a data folder, clear or select the Open on Save option.
- 4. Optionally, if you are the Progeny administrator, then do one or more of the following to modify the security for the individual; otherwise, continue to Step 5.
  - a. Modify the security settings for All Users or for the different classes of users.
    - i. Read? View the data (name and data in the individual datasheet) for the individual.
    - ii. Write? Write data (change the name and enter data to an individual datasheet) to the individual.
  - b. Click Add Class to open the New User Class dialog box to create a new user class, and then set the individual security for the new class.

Figure 3-8: New User Class dialog box



- 5. Click Copy. A message opens indicating that the individual was successfully copied.
- 6. Click OK to close the message and return to the Individuals window.

To move an individual between data folders

Simply drag the selected individual to the appropriate data folder.

# Deleting an Individual

Deleting an individual is a permanent event. After you delete an individual, you cannot recover it from the database. You can delete only singlets from the Individuals window. You must delete individuals who are members of a pedigree from the Pedigree Drawing window. (See To delete an individual from a pedigree)

#### To delete an individual

- 1. On the Progeny main window navigation bar, click the Individuals button that contains the individual that is being deleted.
- 2. Select the individual that is being deleted (CTRL-click to select multiple individuals), and on the main menu, click Individuals → Delete Individual(s).
  - a. A message opens asking you if you are sure that you want to delete the selected individuals click Yes.
- 3. The message closes and the selected individuals are deleted. You return to the Individuals window.

## Moving an Individual to a Pedigree

You can move a singlet (an individual who is not contained in a pedigree) to a pedigree at any time. After you move an individual to a pedigree, the icon for the individual is updated to reflect the fact that the individual is now in a pedigree. You can move an individual to an existing pedigree, or you can create a new pedigree at the time that you are moving the individual, and then move the individual to this new pedigree. When you move an individual to a pedigree, the name that you assigned to the individual is retained for the individual in the pedigree. Also, if the individuals are moved to a pedigrees folder for which the Individual Datasheet template option has been selected, then all the individuals in the folder use the same individual datasheet format, which is extremely useful if you want to manually draw a pedigree for these individuals. (See Manually Drawing a Pedigree)

Figure 3-9: Individual icons



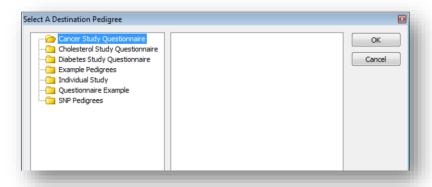
After you move an individual to a pedigree, some options on the Individuals menu are no longer available for the individual such as copying the individual to a data folder, creating a shortcut for the individual, and so on, so make sure that you have carried out all the needed actions for the individual before moving it.

### To move an individual to an existing pedigree



- 1. On the Progeny main window navigation bar, click the Individuals button
- 2. Open the data folder that contains the individual that you are moving to an existing pedigree.
- 3. Select the individual that is being moved, and on the main menu, click Individuals → Move to Pedigree → Existing Pedigree. The Select a Destination Pedigree dialog box opens.

Figure 3-10: Select a Destination Pedigree dialog box

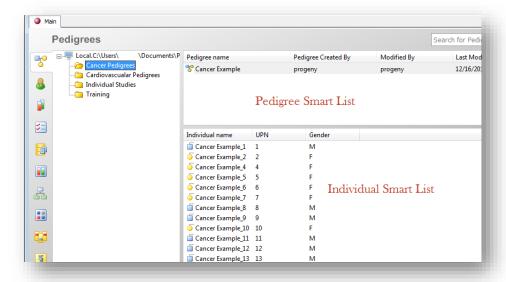


- 4. Select the pedigree to which the individual is being moved, and then click OK.
  - a. The "Select a Destination Pedigree" dialog box closes. You return to the Individuals window and the singlet is removed from the individual data folder.
- 5. Optionally, to view the individual, open the Pedigrees window and in the correct pedigree data folder, select the pedigree to which you moved the individual and you will see this person displayed in the Individuals smart list on the Pedigrees window.



After you add an individual to the pedigree, the icon for the individual is displayed in the upper left corner of the pedigree. You can drag the individual to correct location in the pedigree and manually add the relationships for the individual. See <a href="Manually Drawing a Pedigree">Manually Drawing a Pedigree</a>.

Figure 3-11: Pedigree smart lists

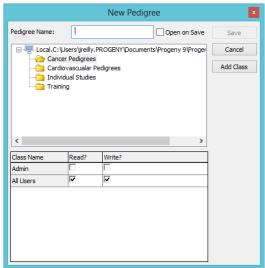


### To move an individual to a new pedigree



- 1. On the Progeny main window navigation bar, click the Individuals button
- 2. Open the data folder that contains the individual that you are moving to a new pedigree.
- 3. Select the individual that is being moved, and on the main menu, click Individuals → Move to Pedigree → New Pedigree. The New Pedigree dialog box opens.

Figure 3-12: New Pedigree dialog box



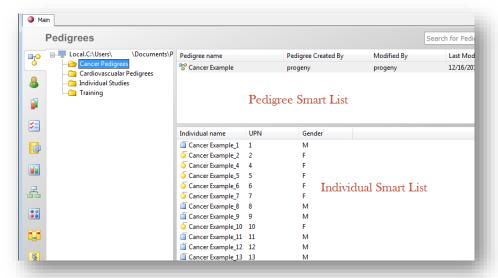
4. Create the new pedigree. See Adding a New Pedigree. Click Save.

- a. A message opens indicating that the new pedigree was successfully added.
- 5. Click OK. The message closes and you are returned to the Individuals window. The singlet is removed from the individual data folder.
- 6. Optionally, to view the individual, open the Pedigrees window and in the correct pedigree data folder, select the pedigree to which you moved the individual. The individual will display in the Individuals smart list on the Pedigrees window.



Because this individual is the first individual that has been added to the pedigree, the individual is automatically designated as the proband. You can always change this designation later.

Figure 3-13: Pedigree Smart Lists





After you add an individual to the pedigree, the icon for the individual is displayed in the upper left corner of the pedigree. You can drag the individual to its correct location in the pedigree and manually add the relationships for the individual. See <a href="Manually Drawing a Pedigree">Manually Drawing a Pedigree</a>.

### Chapter 4 Managing Pedigrees

A pedigree, also known as a genogram, is a diagram that depicts individuals who are related by blood or another factor. Managing pedigrees in Progeny Clinical consists of creating new pedigrees, creating pedigrees shortcuts, creating datasheets for pedigrees, modifying a pedigree, creating pedigrees shortcuts, copying, moving, and deleting pedigrees, merging pedigrees, generating sub-pedigrees, printing pedigrees, and importing pedigree data.

This chapter covers the following topics:

- Adding a New Pedigree
- Formatting a Pedigree Datasheet
- Modifying a Pedigree
- **Creating Pedigree Shortcuts**
- Copying and Moving Pedigrees between Data Folders
- Copying a Pedigree between Databases as a Text File
- Copying a Pedigree between Databases as an XML File
- **Importing FAM Files**
- **Copying and Pasting Pedigrees into Third Party Applications**
- **Merging Pedigrees**
- Deleting a Pedigree
- Generating a Spreadsheet for the Individuals in a Pedigree
- Creating a Sub-Pedigree
- Printing a Pedigree



For detailed information about importing pedigree data, see Appendix B, **Importing Clinical Data** 

### Adding a New Pedigree

When you add a new pedigree, you are essentially adding a blank canvas, or Pedigree Drawing window, for drawing the pedigree. Adding a new pedigree consists of naming the pedigree and selecting the data folder in which to save the pedigrees.

### To add a new pedigree

1. On the Progeny main window navigation bar, click the Pedigrees button.

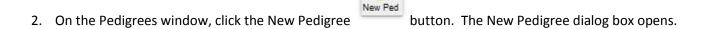
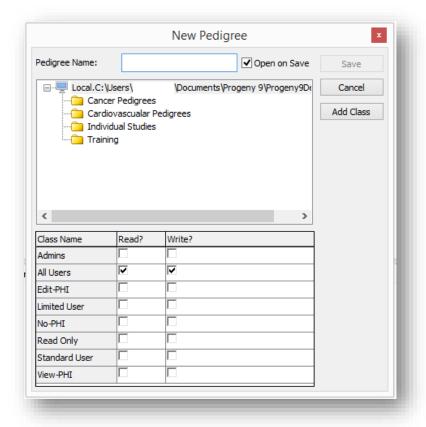


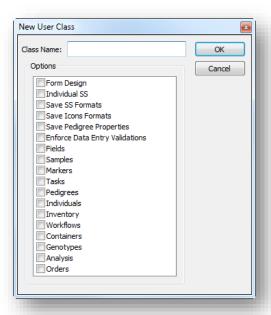
Figure 4-1: New Pedigree Window



3. Enter the name for the new pedigree and choose a folder where it will be added.

- 4. Optionally, click "Open on Save" If you select this option, then after you save the pedigree, a blank pedigree canvas opens in the Pedigree Drawing window. See <a href="Section 2">Section 2</a>, Chapter 5: Drawing and Customizing <a href="Pedigrees">Pedigrees</a>.
- 5. If you are the Progeny administrator, then do one or more of the following to set the security for the pedigree being created; otherwise, continue.
  - a. The security privileges for the Pedigree being created can be set by enabling or disabling the "Read" and "Write" options either for "All Users" within the Progeny database or for the different "user classes" which can be created upon clicking the "Add Class" button shown in the figure above. The "Read" and "Write" functions are defined below:
  - b. Read? View the data (name, data in the pedigree datasheet, the pedigree drawing) for the pedigree.
  - c. Write? Write data (change the name, enter data in the pedigree datasheet, draw the pedigree) to the pedigree.
  - d. Click Add Class to open the New User Class dialog box to create a new user class, and then set the pedigree security for the new class.

Figure 4-2: New User Class window



- 6. Give the class a name and select the desired settings, then click OK.
- 7. A message opens indicating the settings for the new class was added successfully. Click OK to close the message and return to the "New Pedigrees" window.
- 8. Once the new Pedigree has been created with the desired security settings, click Save. The new Pedigree with the set security privileges will be added.

### Formatting a Pedigree Datasheet

You use a pedigree datasheet to enter and store data for a pedigree in a Progeny database. When you create a new pedigree, a pedigree datasheet is automatically created for the pedigree. All the pedigrees that are assigned to a pedigree data folder can use the same datasheet format, or each pedigree in the folder can use a unique datasheet format. You can format a pedigree datasheet at the time that you add a pedigree to a data folder, or later.

#### To format a pedigree datasheet

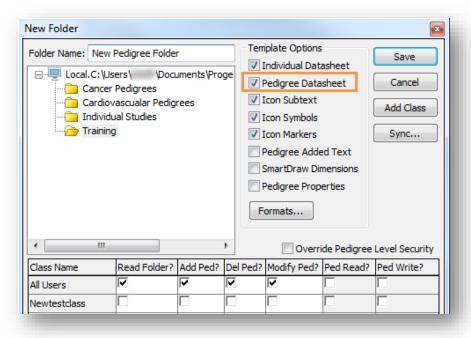
1. Create a pedigree data folder by clicking the New Folder button corner of Main Progeny Window.



found in the upper left

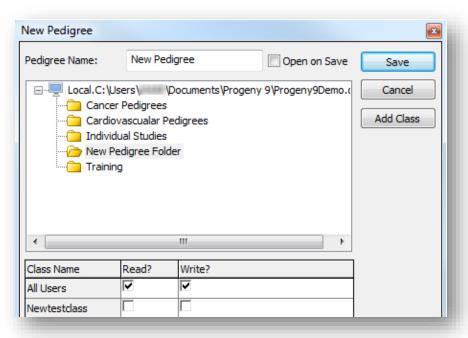
a. If all the pedigrees that are assigned to the folder are to use the same pedigree datasheet format, make sure to select the "Pedigree Datasheet" template option shown in the figure below; otherwise, keep this option unchecked. See <u>Adding Individual and Pedigree Data Folders</u>. After typing in the new Pedigree Folder name and setting the desired Template Options, click "Save".

Figure 4-3: New Pedigree Folder Window



- 2. A message opens indicating the New Pedigree Folder was added successfully, click OK.
- 3. Create a new pedigree by clicking on the New Pedigree button and add the pedigree to the desired data folder (such as "New Pedigree Folder" shown in the figure below).

Figure 4-4: New Pedigree Window



- 4. Once the new Pedigree has been created, click Save. A message opens indicating the New Pedigree was added successfully, click OK.
- 5. Then do one of the following:
  - a. To edit the datasheet of your newly created Pedigree, right-click on the pedigree in the Pedigree window pane, and on the context menu that opens, click "Open Pedigree Datasheet".
  - b. You can click Form Design on this blank datasheet and format the datasheet as needed by utilizing "Style", "Fields", and "Add Text" buttons shown in the figure below.
- 6. If the "Pedigree Datasheet" template option was selected for this Pedigree folder when it was created, then all the pedigrees that you subsequently add to the folder will use the same datasheet format for entering and storing data; otherwise, you must manually format the datasheet for each pedigree that you add to the folder.
- 7. Add all the needed pedigrees to the pedigree data folder. After you have added all the needed pedigrees to the folder, right-click on any pedigree in the folder and on the context menu that opens, click "Open Pedigree Datasheet". If during the creation of this Pedigree's folder, the Pedigree "Datasheet template" option was selected, then all pedigrees that are currently added to the folder or Pedigrees that you continue to add to the folder will use this same datasheet format for entering and storing data; otherwise, you must format a datasheet for each pedigree in the folder.
- 8. See <u>Creating and Formatting Datasheets</u> for detailed information about creating and formatting a pedigree datasheet.

Figure 4-5: Open Pedigree Datasheet

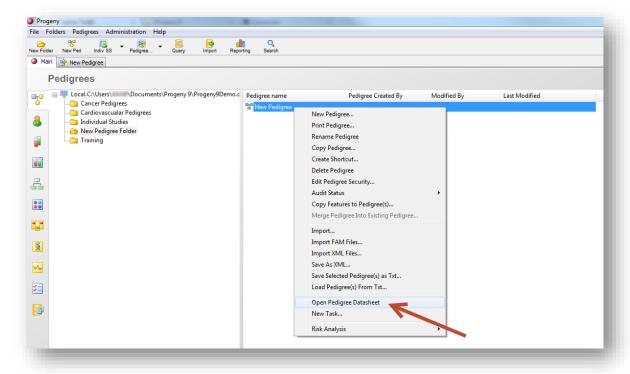
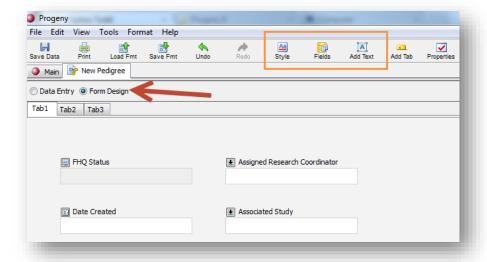


Figure 4-6: Form Design



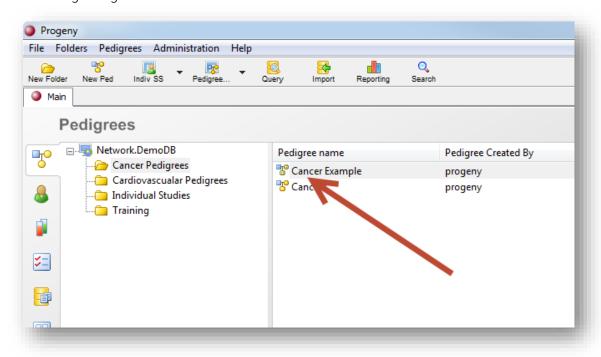
## Modifying a Pedigree

If you are the Progeny administrator, or your administrator has set write access for the pedigree for your user account, you can rename the pedigree. As the Progeny administrator, you can also modify the security levels for the pedigree.

#### To modify a pedigree

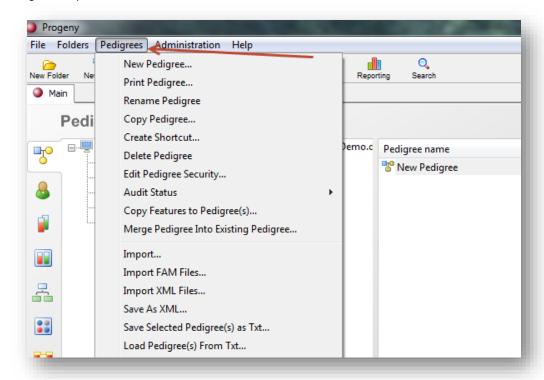
- 1. On the Progeny main window navigation bar, click the Pedigrees button to open the Pedigrees window and choose the folder that has the pedigree you want to modify.
- 2. Open the Pedigree folder that contains the pedigree that is being modified

Figure 4-7: Selecting Pedigree



3. Select the pedigree that is being modified, and on the main menu bar, click Pedigrees, and then click the appropriate option.

Figure 4-8: Pedigrees Option



Option	Description
Rename pedigree	Automatically selects the pedigree name for modification.
Edit Pedigree Security	<ul> <li>Opens the Pedigree Security dialog box in which you can do one or both of the following:</li> <li>Modify the security settings for All Users or for the different classes of users.</li> <li>Read? – View the data (name and data in the pedigree datasheet) for the pedigree.</li> <li>Write? – Write data (change the name and enter data to a pedigree datasheet) to the pedigree.</li> <li>Click Add Class to open the New User Class dialog box to create a new user class, and then set the pedigree security for the new class.</li> </ul>

### **Creating Pedigree Shortcuts**

A pedigree shortcut is a link that points to a pedigree in a Progeny database. You create a pedigree shortcut so that you can add a pedigree to multiple folders without having to copy or duplicate the pedigree. Although a shortcut points to the same original pedigree, each shortcut inherits the template options of the folder in which it is contained. For example, you might place a pedigree shortcut in a Pedigree folder named Diabetes Pedigrees, which has been assigned a pedigree datasheet that has been specifically designed for entering data that is related to the diabetic individuals who are contained in the pedigree. You can also place this same pedigree shortcut in a Pedigree folder named High Blood Pressure Pedigrees, which has been assigned a pedigree datasheet that has been specifically designed for entering data that is related to the hypertensive individuals who are contained in the pedigree.

### To create a pedigree shortcut

- 1. On the Progeny main window navigation bar, click the Pedigrees button to open the Pedigrees window then choose the folder that contains the pedigree for which you are creating the shortcut.
- 2. Select the pedigree for which you are creating the shortcut, and on the main menu, click Pedigrees → Create Shortcut.

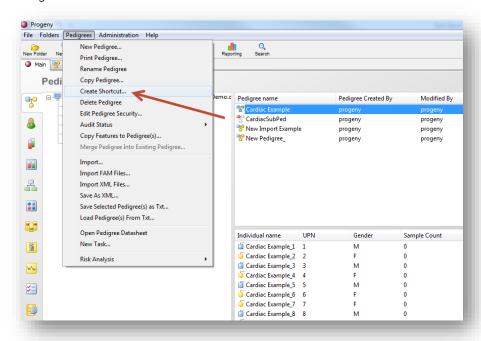
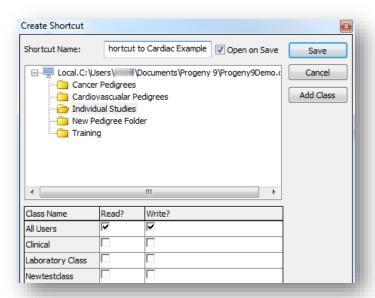


Figure 4-9: Create Pedigree Shortcut

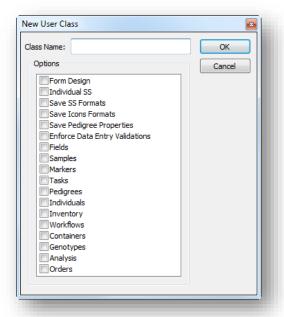
3. The Create Shortcut window opens. The Shortcut Name field is automatically populated with a default value, but you can rename this value.

Figure 4-10: Create Shortcut Window



- 4. Select the folder in which to save the shortcut.
- 5. Optionally, do one or both of the following:
  - a. Change the shortcut name.
  - b. Select "Open on Save".
    - If you select this option, then after you save the pedigree shortcut, the pedigree opens in the Pedigree Drawing window. See <u>Section 2</u>, <u>Chapter 5</u>: <u>Drawing and Customizing</u> <u>Pedigrees</u>.
- 6. If you are the Progeny administrator, then do one or more of the following to set the security for the pedigree; otherwise, continue.
  - a. The security privileges for the Pedigree being created can be set by enabling or disabling the "Read" and "Write" options either for "All Users" within the Progeny database or for the different "user classes" which can be created upon clicking the "Add Class" button shown in the figure above. The "Read" and "Write" functions are defined below:
  - b. Read? —View the data (name, data in the pedigree datasheet, the pedigree drawing) for the pedigree.
  - c. Write? —Write data (change the name, enter data in the pedigree datasheet, draw the pedigree) to the pedigree.
  - d. Click Add Class to open the New User Class dialog box to create a new user class, and then set the pedigree security for the new class.

Figure 4-11: New User Class window



- 7. Give the class a name and select the desired settings, then click OK.
- 8. A message opens indicating the settings for the new class was added successfully. Click OK to close the message and return to the "New Pedigrees" window.
- 9. Once the new Pedigree has been created with the desired security settings, click Save. The new Pedigree with the set security privileges will be added.

### Copying and Moving Pedigrees between Data Folders

You can copy a pedigree between Pedigree folders, and you can move a pedigree between Pedigree folders. For any of these actions, the following are important points to note:

- Copying a pedigree leaves the original pedigree in its original data folder while putting a copy (or duplicate) of the pedigree in the new destination folder. Any edits that you make to the copied pedigree are not reflected in the original pedigree.
- Moving a pedigree removes the pedigree from its original data folder and places the pedigree in a new destination folder. A copy of the original pedigree is not made.

#### To copy or move a pedigree between data folders



open the Pedigrees

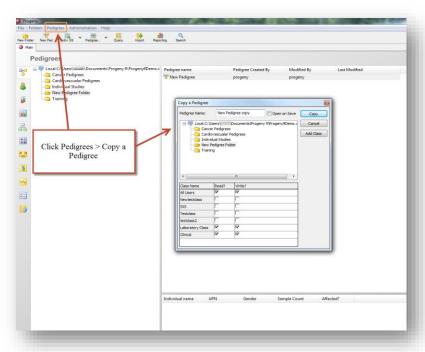
1. On the Progeny main window navigation bar, click the Pedigrees button to window.

2. Open the Pedigree folder that contains the pedigree that you are copying or moving, either copy or move the pedigree to its destination folder using one of the methods outlined in the next sections.

#### To copy a pedigree between Pedigree folders

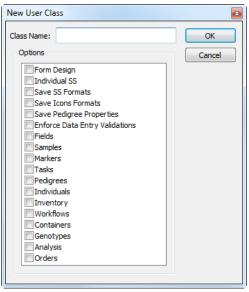
1. On the main menu bar, click Pedigrees > Copy a Pedigree. The Copy a Pedigree window opens.

Figure 4-12: Copy a Pedigree window



- 2. Select the data folder to which the pedigree is being copied.
  - a. Optionally, before you copy the pedigree to a data folder, clear or select the "Open on Save" option.
  - b. Optionally, if you are the Progeny administrator, then do one or more of the following to modify the security for the pedigree; otherwise, continue.
- 3. The security privileges for the Pedigree being created can be set by enabling or disabling the "Read" and "Write" options either for "All Users" within the Progeny database or for the different "user classes" which can be created upon clicking the "Add Class" button shown in the figure above. The "Read" and "Write" functions are defined below:
  - a. Read? View the data (name, data in the pedigree datasheet, the pedigree drawing) for the pedigree.
  - b. Write? Write data (change the name, enter data in the pedigree datasheet, draw the pedigree) to the pedigree.
- 4. Click "Add Class" to open the New User Class window to create a new user class, and then set the pedigree security for the new class.

Figure 4-13: New User Class window



- 5. Give the class a name and select the desired settings, then click OK.
- 6. A message opens indicating the settings for the new class was added successfully.
- 7. Click OK to close the message and return to the "Copy a Pedigree window".
- 8. Once your copied pedigree has been given a name, click "Copy". At this point, if you select the Pedigree folder that the copied pedigree was designated for, you will see that the copy now resides in that folder.

#### To move a pedigree between Pedigree folders

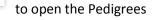
Simply drag the selected pedigree to the appropriate Pedigree folder.

### Copying a Pedigree between Databases as a Text File

When you copy a pedigree between databases as a text file, you can copy pedigree data, individual data, sample data, and markers.

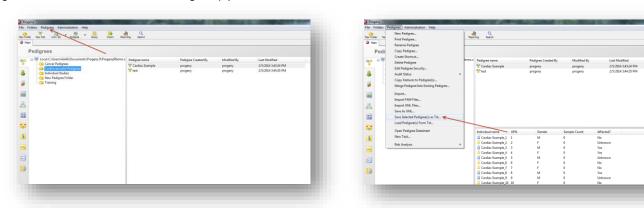
#### To copy a pedigree between databases as a text file

- 1. Launch Progeny and log into the database from which you are coping the pedigrees.
- 2. On the Progeny main window navigation bar, click the Pedigrees button window and choose the folder that has the pedigree you want to copy.



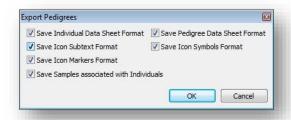
3. Select the pedigree that you are copying, (CTRL-click to select multiple pedigrees) and on the main menu, click Pedigrees → "Save Selected Pedigree(s) as Txt".

Figure 4-14: Save Selected Pedigree(s) as Txt



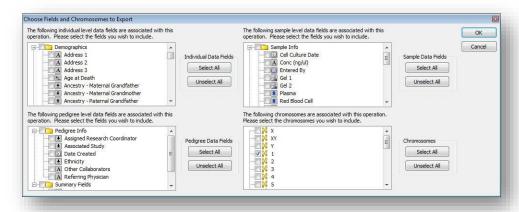
4. The Export Pedigrees dialog box opens. This dialog box lists all the options that can be exported with the pedigrees. By default, all options are selected.

Figure 4-15: Export Pedigrees window



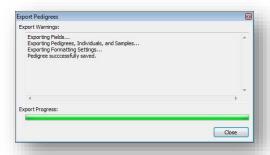
- 5. Optionally, clear any options that are not be exported with the pedigree(s), and then click OK.
- 6. The Export Pedigrees window closes and the Choose Fields and Chromosome to Export window opens.

Figure 4-16: Choose Fields and Chromosome to the Export dialog box



- 7. Select the data fields and chromosomes (if chromosome data is present) that are to be exported with the pedigrees, and then click OK.
- 8. Enter a name for the exported pedigree(s) file (the file type is set to .txt and you cannot change this), browse to the appropriate location that your exported file will be saved, and then click "Save".
- 9. An Export Pedigrees dialog box opens, indicating the status of the export.

Figure 4-17: Export Pedigrees window

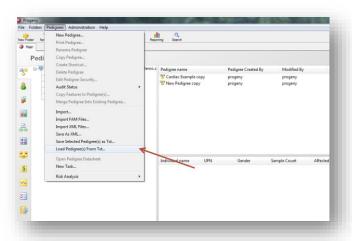


- 10. When the export is complete, click Close.
- 11. On the Progeny main menu, click File > Exit to log out of the database from which you are copying the pedigrees, and then continue.

#### To load a pedigree text file

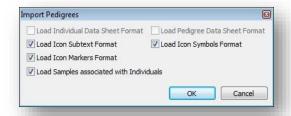
- 1. Launch Progeny and log into the database to which you are copying the pedigrees.
- 2. On the Progeny main window navigation bar, click the Pedigrees button window and choose the folder you want to load the pedigree into.
- to open the Pedigrees
- 3. On the main menu, click Pedigrees → Load Pedigree(s) from Txt.

Figure 4-18: Load Pedigree(s) From Txt



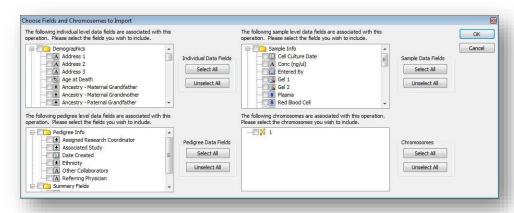
- 4. The Open file window appears. Locate the pedigree .txt file that you are loading, and then click Open.
- 5. The Import Pedigrees dialog box opens. This dialog box lists all the options that were imported with the selected pedigree.

Figure 4-19: Import Pedigrees dialog box



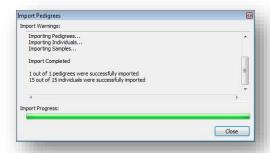
- 6. You can leave the options as-is or clear the options that are not to be imported with the pedigree. Click OK.
- 7. The Choose Fields and Chromosome to Import dialog box opens. This dialog box lists all the data fields and chromosomes that are associated with the imported pedigree.

Figure 4-20: Choose Fields and Chromosomes to Import dialog box



- 8. Select the data fields and chromosomes (if chromosome data is present), that are to be imported with the pedigree and then click OK.
- 9. The Import Pedigrees dialog box opens, indicating the status of the import.

Figure 4-21: Import Pedigrees dialog box

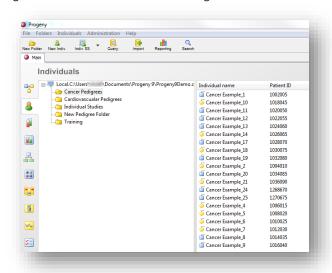


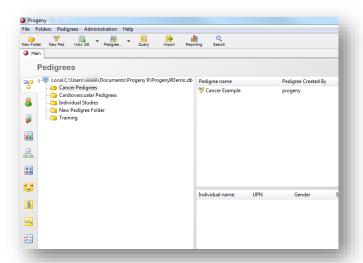
- 10. When the import is complete, click Close.
- 11. Individuals in the pedigree are imported into a folder that is named identically to the folder into which you imported the pedigrees. (Remember, when you create a new individual data folder, the same folder will be created as a pedigree folder with the same template options.
  - a. Likewise, when you create a new Pedigree folder, the very same folder will be created as an individual data folder with the same template options.



Both individuals and Pedigrees have the exact same folders; if a pedigree folder is deleted the individual data folder will also be deleted.

Figure 4-22: Individual and Pedigree Folders





12. The folder structure for the imported database fields is recreated in the database into which you are importing the pedigrees, and the database fields are imported into the appropriate folders.

### Copying a Pedigree between Databases as an XML File

When you copy a pedigree between databases as a .xml file, you can copy pedigree data, individual data, and markers. You cannot copy sample data. Also, to be able to select the data fields and markers that are to be copied (versus having all data fields and markers copied by default), you must use the Save as XML and Load From XML options that are available on the context menu from an open pedigree, and not the options that are available from the main menu.

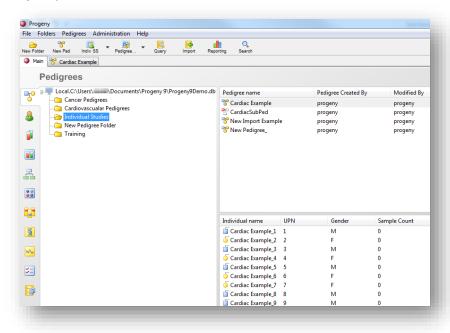
#### To copy a pedigree between databases as an XML file

- 1. Launch Progeny and log into the database from which you are coping the pedigree.
- 2. On the Progeny main window navigation bar, click the Pedigrees button window then choose the folder that has the pedigree you wish to copy.



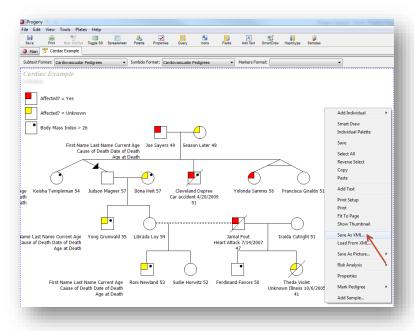
to open the Pedigrees

Figure 4-23: Select Pedigree folder



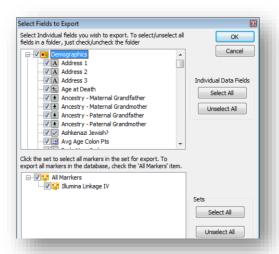
3. Open the pedigree that you are saving as a .xml file, right-click in the Pedigree Drawing window, and on the context menu that opens, click "Save As XML" (See figure below).

Figure 4-24: Save As XML



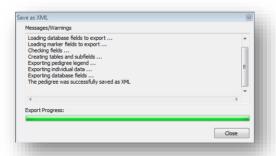
- 4. The "Save As" dialog box opens.
- 5. Enter a name for the exported pedigree(s) file (the file type is set to .xml and you cannot change this), browse to the appropriate location that your exported file will be saved, and then click "Save".
- 6. The Select Fields to Export dialog box opens. This dialog box lists all the pedigree data fields, individual data fields, and markers that are available for exporting for the pedigree. By default, all data fields and markers are selected for export.

Figure 4-25: Select Fields to Export dialog box



7. Optionally, clear the selections for the data fields and/or markers that are not to be exported, and then click OK. The Save as XML dialog box opens, indicating the status of the export.

Figure 4-26: Save as XML dialog box

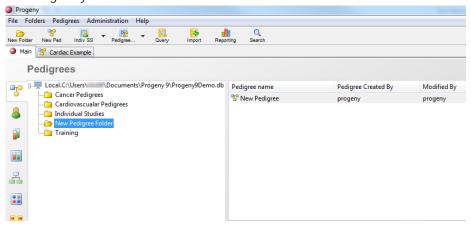


- 8. When the export is complete, click Close.
- 9. On the Progeny main menu, click File > Exit to log out of the database from which you are copying the pedigrees, and then continue.

#### To load a pedigree XML file

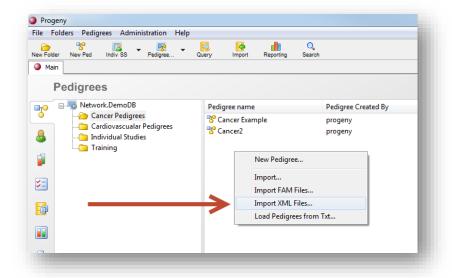
- 1. Launch Progeny and log into the database to which you are copying the pedigrees.
- 2. On the Progeny main window navigation bar, click the Pedigrees button to open the Pedigrees window and choose the folder where you want to load the XML file into.

Figure 4-27: Select Pedigree folder



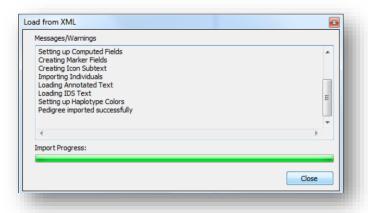
3. Right-click on the Pedigree pane for the selected folder, and on the context menu that opens, click "Load From XML".

Figure 4-28: Load From XML



- 4. A window opens asking you to choose the folder where your XML file(s) is/are stored.
  - a. NOTE: This process will import all XML files from the selected folder

Figure 4-29: Load from XML dialog box



5. After the XML file is imported, click Close.

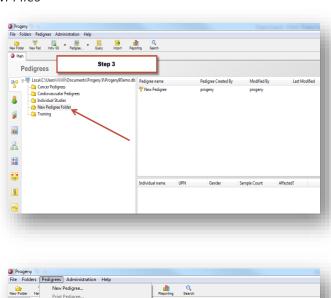
# **Importing FAM Files**

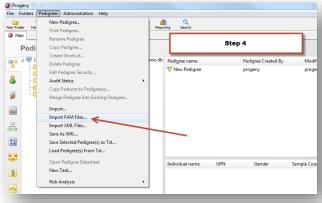
You can import multiple Cyrillic pedigrees (FAM files) into a Progeny database.

### To import FAM files

- 1. Launch Progeny and log into the database into which you are coping the pedigree.
- 2. On the Progeny main window navigation bar, click the Pedigrees button window.
- to open the Pedigrees
- 3. Select the Pedigree folder into which you are copying the pedigrees (See example in the figure below).
- 4. On the main menu, click Pedigrees → Import FAM Files.

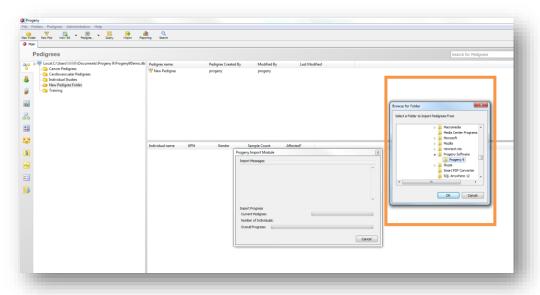
Figure 4-30, 4-31: Import FAM Files





5. The Browse for Folder dialog box opens. Browse to the location that contains the FAM files you desire to import, and then click OK.

Figure 4-32: Location of FAM File



6. A Progeny Import Module dialog box opens, indicating the status of the import. When the import is complete, click Close.

# Copying and Pasting Pedigrees into Third Party Applications

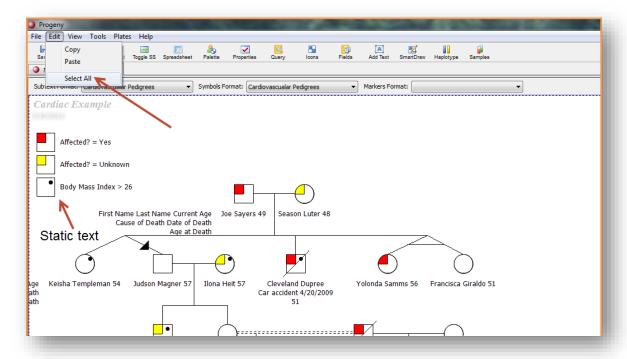
Progeny enables you to copy and paste the image of your pedigree from the Pedigree Drawing window into a third-party application such as Microsoft Word or Microsoft PowerPoint. When you are copying and pasting a pedigree into a third-party application, note the following:

- Ideally, you should have the pedigree formatted and drawn to the appropriate specifications before you copy it.
- Any icon subtext or haplotypes that are displayed on the pedigree are also copied; however, any static text that is displayed on the pedigree is not copied. If necessary, you can copy this text separately.

#### To copy and paste a pedigree into a third-party application

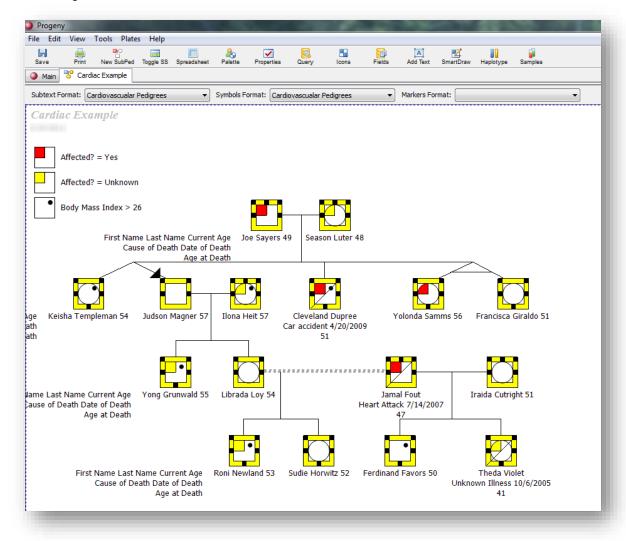
1. Open the pedigree that is being copied so that the Pedigree Drawing window is displayed, click Edit → Select All.

Figure 4-37: Edit menu



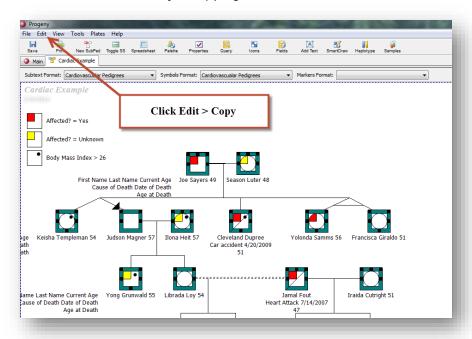
2. Every individual icon in the pedigree has become a double yellow square with handles on it, indicating that is has been selected.

Figure 4-38: Pedigree with all icons selected



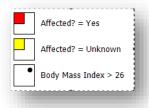
- 3. On the main menu, click Edit  $\rightarrow$  Copy.
- 4. Also, to accomplish the same task you can:
  - a. Use the standard keyboard copy command of CTRL + C.
  - b. Right-click anywhere in the Pedigree Drawing window, and on the context menu that opens, click Copy.
- 5. Regardless of which route you take every individual icon in the pedigree will become a double green square with handles on it, indicating that is has been selected for copying.

Figure 4-39: Pedigree with all icons selected for copying



- 6. Open a blank Word document or PowerPoint slide and use the standard menu or keyboard commands to paste the pedigree.
- 7. After pasting the pedigree:
  - To copy the static text to the pedigree, click on the static text in the pedigree to select it. (The static text is displayed in a text box with handles and the cursor changes to a set of double-headed arrows.)
  - Right-click on the selected text and select Copy or use the standard keyboard copy command of CTRL + C to copy the selected text, and then use the standard menu or keyboard commands to paste the text into the third-party application containing pedigree.

Figure 4-40: Selected static text on a pedigree



## **Merging Pedigrees**

If you have two pedigrees that are in the same database and all the individuals in both pedigrees are from the same family, you can merge the pedigrees. When you merge pedigrees, all the data that is associated with everyone in the pedigree remains intact.



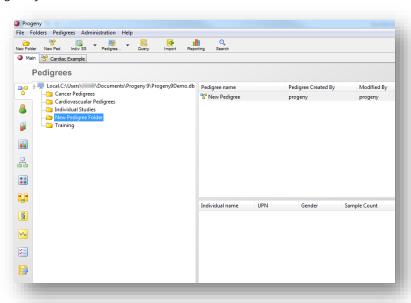
Keep in mind that merging a Pedigree is a permanent event and once executed cannot be undone. You can merge only original pedigrees. You cannot merge copies of original pedigrees.

### To merge pedigrees



- 1. On the Progeny main window navigation bar, click the Pedigrees button window.
- to open the Pedigrees
- 2. Open the Pedigree folder that contains the pedigree that is being merged into another pedigree.

Figure 4-41: Select Pedigree folder



3. Select the pedigree that is being merged into another pedigree, and on the main menu, click Pedigrees → Merge Pedigree into Existing Pedigree.

Figure 4-42: Merge Pedigree into Existing Pedigree

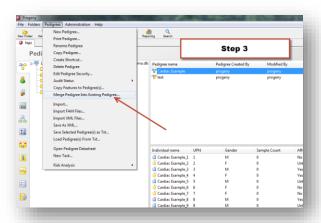
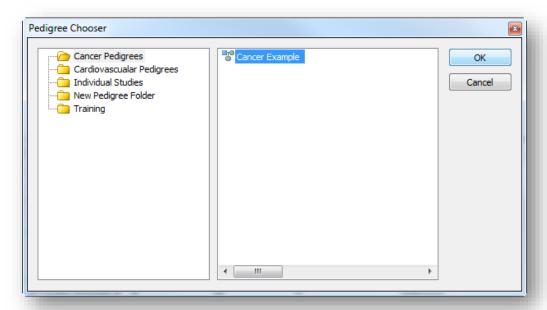


Figure 4-43: Pedigree Chooser dialog box



- 4. Select the pedigree into which you are merging another pedigree, and then click OK.
- 5. A message opens indicating that the pedigrees were successfully merged. Click OK to close the message and return to the Pedigrees window.
- 6. The entry for the pedigree that was merged is no longer displayed in the window. Only the entry for the pedigree into which the merge occurred is displayed. For example, if you merged a pedigree named Patient 102 into a pedigree named Patient 110, then the entry for Patient 102 is no longer displayed in the Pedigrees window. Only the entry for Patient 110 is displayed. Once again, please understand that merging a Pedigree is a permanent event and once executed cannot be undone.

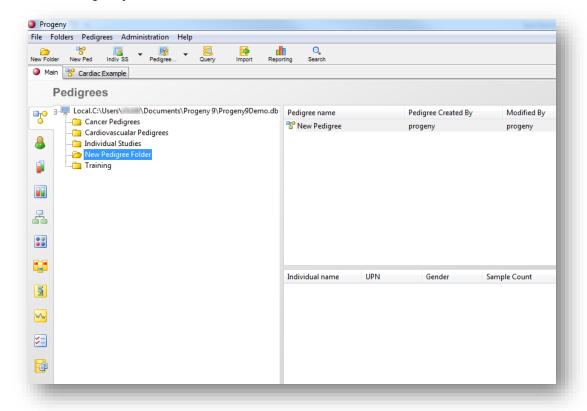
## Deleting a Pedigree

Deleting a pedigree is a permanent event. After you delete a pedigree, you cannot recover it from the database.

#### To delete a pedigree

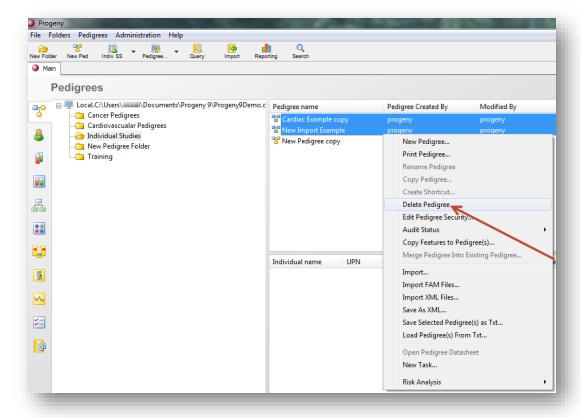
1. On the Progeny main window navigation bar, click the Pedigrees button to open the Pedigrees window then choose the Pedigree folder that contains the pedigree(s) you want to delete.

Figure 4-44: Select Pedigree folder



2. Select the pedigree that is being deleted (CTRL-click to select multiple pedigrees), and right-click on Pedigree(s) being copied, on the context menu that opens click Delete Pedigree.

Figure 4-45: Delete Pedigree



- 3. A message opens asking you if you are sure that you want to delete the selected pedigrees.
- 4. Click Yes. The message closes and the selected pedigrees are deleted. You return to the Pedigrees window.

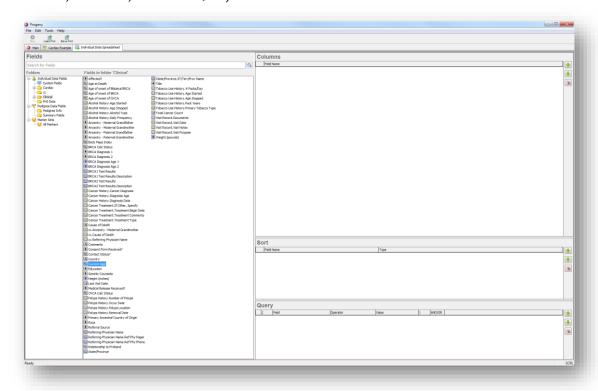
# Generating a Spreadsheet for the Individuals in a Pedigree

<u>Section 1, Chapter 4: Spreadsheet Basics</u> contains a much greater level of detail about the operation of the spreadsheet module. You can also generate a spreadsheet that consists solely of the individuals in a selected pedigree.

#### To generate a spreadsheet for the individuals in a pedigree

- 1. Open the appropriate pedigree in the Pedigree Drawing window.
- 2. On the window's toolbar, click the Spreadsheet button Spreadsheet Format window. Spreadsheet Format window.

Figure 4-45: Fields, Columns, Sort and Query window



3. From the "Fields" window pane, drag all desired fields (including any system fields) that you want to be displayed in the spreadsheet onto the "Columns" window pane (Please reference figure below).

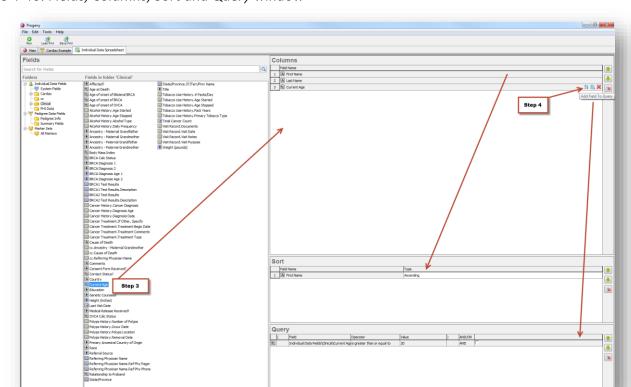
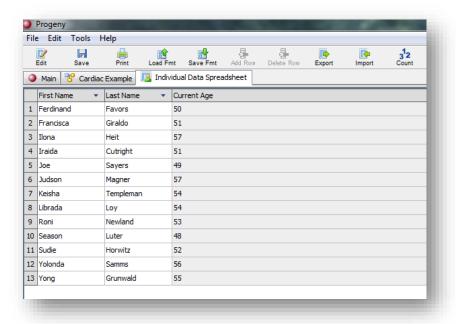


Figure 4-46: Fields, Columns, Sort and Query window

- 4. Once all desired fields are within the "Columns" window pane, hovering your mouse over any of the fields will display buttons on the far-right side with options to Add Field to Sort or Add Field to Query as shown in the figure above:
  - a. Sort: The data in the spreadsheet will be sorted (ordered) by the field of your choosing.
  - b. Example: The "First Name" field was added to the Sort window pane. Under the "Type" column to the right of this field, there is a drop-down box that allows you to order the rows within your spreadsheet by two options: Ascending or Descending order. With this field set to be sorted in ascending order the spreadsheet will be organized by the first name in alphabetical order.
  - c. Query: This will allow you to filter the results for all fields within your spreadsheet based on the field(s) added to the "Query" window pane.
  - d. Example: The "Current Age" field was added to the Query window pane; within the "Operator" column it is set when the spreadsheet is run, to only show ages "greater than or equal to". In the Value column, which is the next column over, the numeric value of "20" is typed. With these settings in place, the spreadsheet will be set to only show the data of patients age "20" and over.

- 5. Once the format for the spreadsheet has been finalized proceed to click the Run button in the upper left corner of Progeny window to generate the spreadsheet.
- 6. All the rules and caveats for entering data into a spreadsheet, loading and saving formats, running queries, and so on still apply to a spreadsheet that was just generated for individuals in a selected pedigree. See Section 1, Chapter 4: Spreadsheet Basics.

Figure 4-46: Spreadsheet



7. Once you have the desired spreadsheet results click the "Save Fmt" button and specify a name and save the spreadsheet. This and other spreadsheets can be loaded for future use by clicking on the Load Fmt



button.

# Creating a Sub-Pedigree

A sub-pedigree is simply a subset of individuals from an existing pedigree. The data that is associated with everyone in a sub-pedigree is the actual data for the individual. To view the pedigree with selected members hidden, you would simply create a sub-pedigree and save it for later retrieval. For instance, you might want to save a subset of a pedigree in which only the first-degree relatives of the proband are displayed. You simply create a subset of the pedigree where these are the only individuals shown on the pedigree. Some important points to note about a sub-pedigree are the following:

- There are no limits as to the number of sub-pedigrees that you can create for a given pedigree.
- You can add individuals to a pedigree only from within the pedigree, and not from within any sub-pedigrees.
- Any changes that you make to the structure or data in the pedigree are reflected in the associated subpedigrees as well. For instance, if you delete the proband from the pedigree, the proband is also deleted from any associated sub-pedigrees.
- Any changes that you make to the structure of a sub-pedigree are not reflected in the pedigree. For
  instance, if you delete the proband from a sub-pedigree, the proband is not deleted in the pedigree.
- Any changes that you make to the data for an individual in a sub-pedigree are reflected in the pedigree.
   (Remember, the data that is associated with everyone in a sub-pedigree is the actual data for the individual.)
- You can open the individual datasheet for an individual in a sub-pedigree the same way that you open it for an individual in a pedigree. Simply right-click on the individual, and on the context menu that opens, click Open Individual Datasheet.
- You can generate a spreadsheet for all the individuals who are in a sub-pedigree in the same way you
  generate a spreadsheet for all the individuals in a pedigree. Open the sub-pedigree, and on the Pedigree
  Drawing window toolbar, click the Spreadsheet button. (See <u>To generate a spreadsheet for the individuals in
  a pedigree</u>)

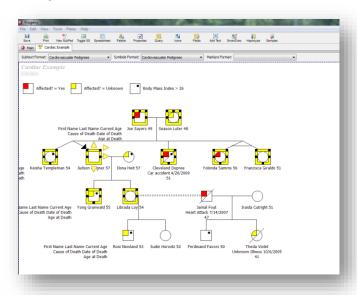
### To create a sub-pedigree



The following procedure details the creation of a sub-pedigree for all 1st-degree relatives of a proband in a pedigree; however, by analogy, you can follow this procedure to create any sub-pedigree as needed.

- 1. Open the pedigree for which you are creating a sub-pedigree in the Pedigree Drawing window.
- 2. Right-click on the proband, and on the context menu that opens, click Select → 1st Degree. All individuals who are first degree relatives to the proband are selected in the pedigree.

Figure 4-47: Sub-Pedigree First Degree Selection

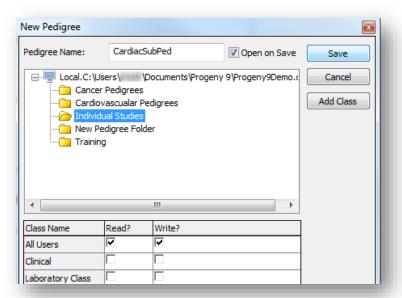


3. On the Pedigree Drawing window toolbar, click the New Sub-Pedigree Pedigree dialog box opens.



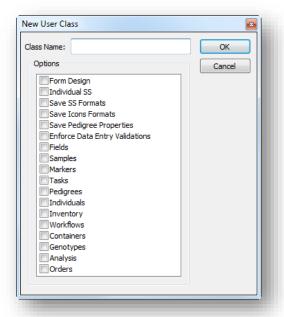
button. The New

Figure 4-48: New Sub-Pedigree window



- 4. Enter the name of the new sub-pedigree and choose the folder where it will be added to.
- 5. Optionally, click Open on Save. If you select this option, then after you save the pedigree, the sub-pedigree opens in the Pedigree Drawing window. See Section 2, Chapter 5: Drawing and Customizing Pedigrees.
- 6. If you are the Progeny administrator, then do one or more of the following to set the security for the subpedigree; otherwise, continue.
  - a. The security privileges for the New Sub-Pedigree being created can be set by enabling or disabling the "Read" and "Write" options either for "All Users" within the Progeny database or for the different "user classes" which can be created upon clicking the "Add Class" button shown in the figure above. The "Read" and "Write" functions are defined below:
  - b. Read? View the data (name, data in the pedigree datasheet, the pedigree drawing) for the pedigree.
  - c. Write? Write data (change the name, enter data in the pedigree datasheet, draw the pedigree) to the pedigree.
  - d. Click Add Class to open the New User Class dialog box to create a new user class, and then set the pedigree security for the new class.

Figure 4-49: New User class window



- 7. Give the class a name and select the desired settings, then click OK.
- 8. A message opens indicating the settings for the new class was added successfully. Click OK to close the message and return to the "New Pedigree" window.
- 9. Once the new Pedigree has been created with the desired security settings click Save then click OK to close the message and open the sub-pedigree in the Pedigrees Drawing window.

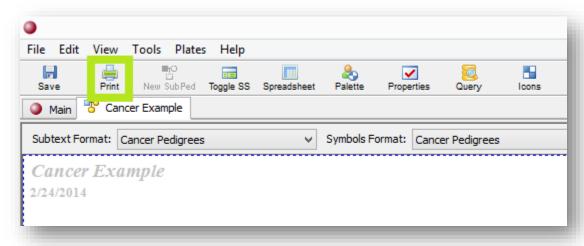
# Printing a Pedigree

You can print a pedigree that is displayed in the Pedigree Drawing window.

### To print a pedigree

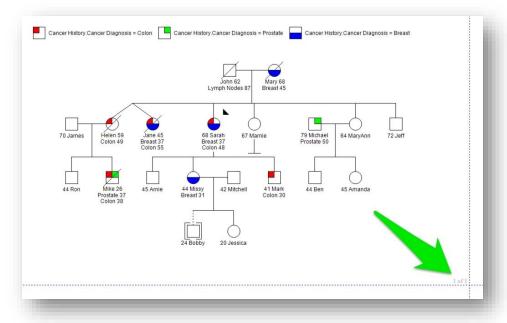
1. Open the pedigree you want to print then click the Print button Print in the top-left corner of the icon menu.

Figure 4-50: The print button in the icon menu



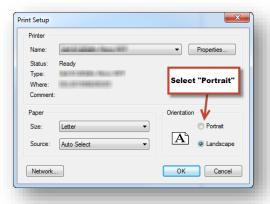
- 2. You can optionally set one or more of the following options:
  - a. Turn on the Show Page Breaks option within the Progeny tab of the Properties dialog box. (See <u>Properties dialog box</u>, <u>Progeny tab</u>) If this option is selected, blue lines will be displayed on the pedigree viewer to show how your pedigree will print. Page numbers are displayed in the bottom right corner of each page in the order of how the pages print (This can be observed below).

Figure 4-50: Pedigree Drawing window



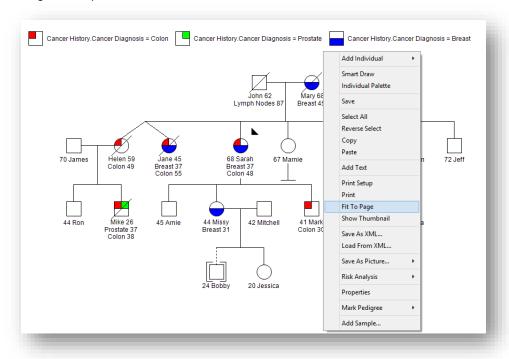
b. Landscape is the default mode for printing. To change this, on the main menu, in the upper left-hand corner, click File > Printer Setup, and on the Print Setup dialog box, select Portrait.

Figure 4-51: Print Setup



c. To automatically scale the pedigree to print on a single page, right-click on the opened pedigree and choose Fit to Page. To return the pedigree to its original scale, enter the appropriate value for the Pedigree Scale option on the Pedigree tab of the Properties dialog box. See <a href="Properties dialog box">Properties dialog box</a>, <a href="Pedigree tab">Pedigree tab</a>.

Figure 4-52: Fit to Page Example



- On the pedigree toolbar in the upper left corner, click the Print button
   The Print dialog box opens.
- 4. On the Print dialog box, select the needed printing options, and then click Print.

# Chapter 5 – Drawing and Customizing Pedigrees

A pedigree, also known as a genogram, is a diagram that depicts individuals who are related by blood or another factor. When you create a new pedigree, you are essentially creating a blank canvas for drawing the pedigree. (This blank canvas is referred to as the Pedigree Drawing window.) Drawing of a pedigree consists of identifying the icons that are to be used in the pedigree, including their subtext and symbols, adding individuals to the pedigree, deleting individuals from a pedigree, adding and modifying relationships among individuals in the pedigree, and so on.

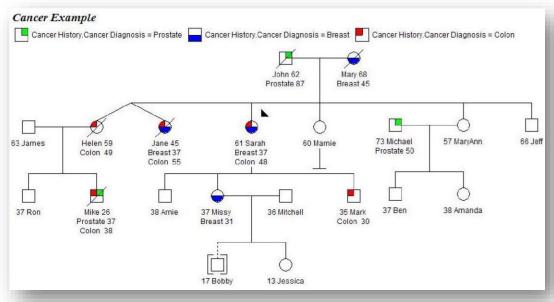
This chapter covers the following topics:

- Overview of a Pedigree
- Overview of Pedigree Icons
- <u>Pedigree Properties</u>
- Configuring Pedigree Icons
- Saving and Loading Icon Formats
- Manually Drawing a Pedigree
- Working with Individuals in a Pedigree
- Adding an Individual Shadow to a Pedigree
- Creating a Pedigree Thumbnail

## Overview of a Pedigree

A pedigree, also known as a genogram, is a diagram that depicts individuals who are related by blood or another factor. Different icons represent different types of individuals. For example, a square icon indicates a male individual, a circle icon indicates a female individual, and a diamond icon indicates an individual of unknown/undetermined gender. Symbols are applied to icons to graphically represent data about the individual, such as individuals with a cancer diagnosis that equals Breast. A legend identifies each symbol. The lines connecting individuals identify the relationship between two individuals, such as marital status. The icon subtext, which is the text that is displayed above or below an individual icon on a pedigree, is specific for the individual. Figure 5-1 below shows a pedigree that details the cancer history for the members of a family.

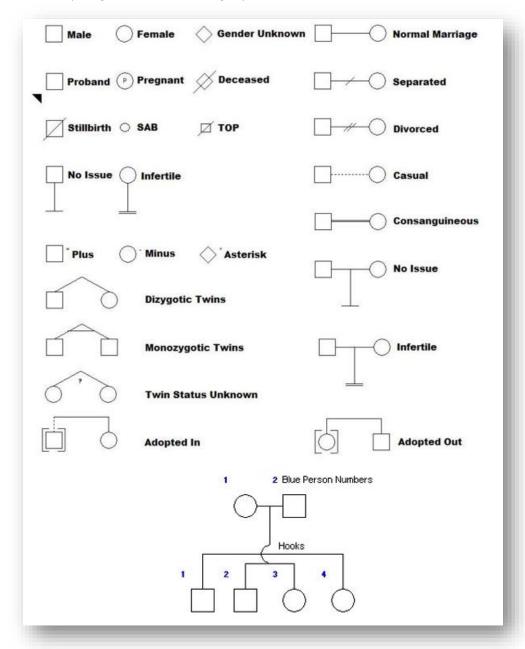
Figure 5-1: Example of a pedigree



# Overview of Pedigree Icons

Although there are no definitive standards when drawing pedigrees, there are published guidelines for the human genetics market. Progeny uses pedigree icons based on the guidelines published in the American Journal of Human Genetics (1995) Bennet, et al.

Figure 5-2: Icons used in pedigrees drawn in Progeny



# **Pedigree Properties**

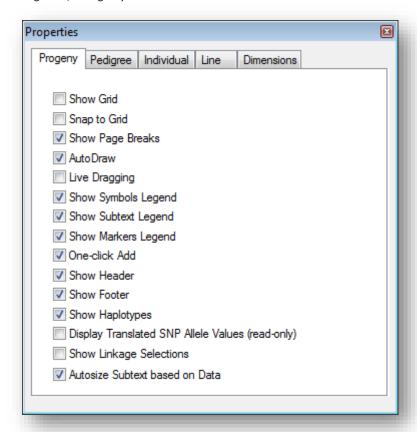
The Properties dialog box displays all the options that you can set for controlling the drawing and displaying of a pedigree. Although some of these options might not make sense right now, it is important for you to be aware of them so that when they are referenced later in this chapter, you are able to correctly modify and apply the properties when

you are drawing pedigrees. You open the Properties dialog box by clicking on the Properties button Properties on the toolbar for any open pedigree.

The dialog box has five tabs—Progeny, Pedigree, Individual, Line, and Dimensions. By default, the Progeny tab is the active tab when the dialog box first opens.

#### Properties dialog box, Progeny tab

Figure 5-3: Properties dialog box, Progeny tab

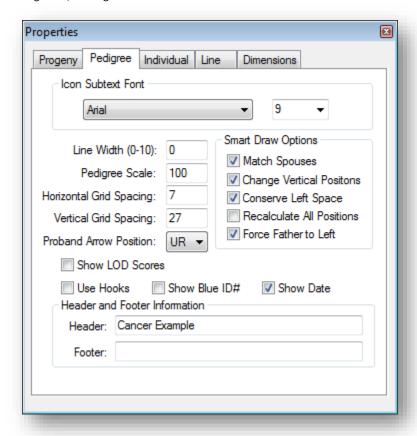


# Description of Properties → Progeny Tab options

Option	Description
Show Grid	Displays a grid on the pedigree canvas.
Snap to Grid	When moving an icon in the pedigree, aligns (or "snaps") the icon to the nearest intersection of lines in the grid, even if the grid is not visible.
Show Page Breaks	Displays print boundaries on the pedigree as blue dashed lines.
AutoDraw	Automatically runs SmartDraw every time you add an individual to a pedigree. You can then define your SmartDraw options. See <a href="Smart Draw Options">Smart Draw Options</a> .
Live Dragging	Relationship lines move with the icon when you reposition it.
Show Symbols Legend	Displays a legend for the symbols shown on the pedigree.
Show Subtext Legend	Displays a legend for the icon subtext displayed in the pedigree.
Shoe Markers Legend	Displays a legend for the icon markers displayed in the pedigree.
One-click Add	Allows you to click on specific handles on an individual icon to automatically add a relationship to the individual. (You do not have to manually add relationships from the palette or drag lines between icons to create relationships).
Show Header/Show Footer	Displays a header or footer on each page of the pedigree. Customizable from Pedigree Tab of Properties menu.
Show Haplotypes	Display a colored-coded legend for markers that are displayed on a pedigree.
Display Translated SNP Allele Values	Displays the SNP allele values next to the individual's icon.
Show Linkage Selections	Displays sample status (Sample/Plate/Genotype) on the pedigree and indicates whether an individual has been "Included in Analysis."
Autosize Subtext based on Data	Dynamically resizes the icon subtext based on the quantity of text entered for an individual.

## Properties dialog box, Pedigree tab

Figure 5-4: Properties dialog box, Pedigree tab



# Description of Properties → Pedigree Tab options

Option	Description
Icon Subtext Font	The font type and font size used for the icon subtext.
Line Width	The width of the relationship lines used in the pedigree.
Pedigree Scale	Size of the pedigree, from 0 to 800%. 100 is the default – values below this will shrink the pedigree while values larger will increase the size.
Grid Spacing Horizontal/Vertical	If Show Grid is selected on the Progeny tab, then change these values to adjust the spacing between the grid lines – measured in pixels.
Proband Arrow Position	Change the position of the solid black arrowhead that identifies the proband on a pedigree. Values are UL (for upper left), UR (for upper right), LL (for lower left), and LR (for lower right).
	Match Spouses – Keep spouse pairs as close to each other as possible.
	Change Vertical Positions – Aligns all individuals of the same generation along with a single horizontal plane.
Smart Draw Options	Converse Left Space – Moves the pedigree as far to the left as possible.
Smart Braw Options	Recalculate All Positions – Forces Smart Draw to disregard the existing positions of individuals when redrawing the pedigree.
	Force Father to the Left – The Male member of a spouse pair will <u>always</u> be positioned to the left of the Female.
Show LOD Scores	Displays a table with LOD scores for the markers that are listed for everyone.
Hea Haaks	Distinguishes between lines that cross over one another.
Use Hooks	Note: See Overview of Pedigree Icons for a graphical depiction of hooks.
Show Blue ID #	Display a blue number next to an icon in a pedigree to indicate the ordering of individuals from left to right in a generation.
	Note: See Overview of Pedigree Icons for a graphical depiction of blue numbers. You can change the position of blue numbers on the Dimensions tab.
Show Date	Display the current date in the upper left-hand corner of the pedigree.
Header and Footer Information	Optional text-entry fields for header and footer information.

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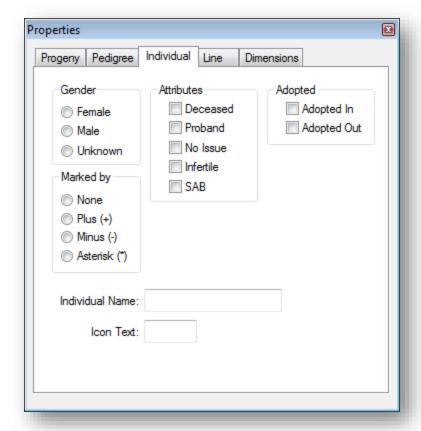
### Properties dialog box, Individual tab

This tab is used to change the properties of an individual. The options here will have no effect on the pedigree unless an individual is selected. To modify an individual's properties via this tab, select the icon for that individual (the selected icon becomes a double yellow square with handles on it), and then open the Individual tab on the Properties dialog box, and change the values for all options as needed.

Figure 5-5: Selected icon and relationship attachment points



Figure 5-6: Properties dialog box, Individual tab



#### Description of Properties → Individual Tab options

Option	Description
Gender	Change the gender of the selected individual.
Attributes	Select the attributes for the selected individual.
Adopted	Indicate the adoption status for the selected individual.
Marked By	Place the indicated symbol next to the selected individual.
Individual Name	Every individual in Progeny is assigned an individual name (a system field). When you are drawing a pedigree in Progeny the software automatically assigns an individual name to an individual. The default format for the Individual Name is pedigree name _UPN. If needed, you can edit this default name here.
Icon Text	Enter in the text that is to be displayed inside the individual icon. The text can be any number of alphanumeric characters or special characters.

## Properties dialog box, Line tab

You use the options on the Line tab to change the properties of a relationship line. Select the line on the pedigree for which you are changing the properties, (a black dot is displayed at each end of a selected line), then open the Line tab on the Properties dialog box and change the values for all options as needed.

Figure 5-7: Example of a selected line in a pedigree

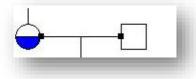
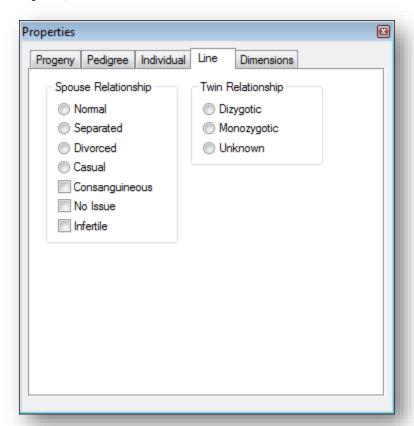


Figure 5-8: Properties dialog box, Line tab

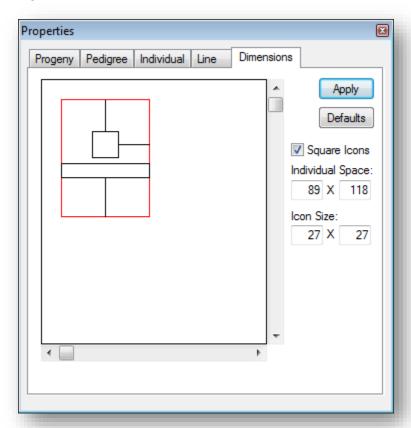


Option	Description
Spouse Relationship	Applicable only to a selected spouse relationship line. The displayed line indicates the relationship between the spouses.
Twin Relationship	Applicable only to a selected twin relationship line. The displayed line indicates the relationship between the twins.

### Properties dialog box, Dimensions tab

You use the options on the Dimensions tab to change the dimensions (Individual Space or Icon Size) of a pedigree.

Figure 5-9: Properties dialog box, Dimensions tab



Option	Description
Individual Space	The space between individual icons in a pedigree.
Icon Size	The size of the icons that represent individuals in a pedigree. The Square Icons option locks icon height and width to identical values.

Note: You can also manually adjust the individual space and icon size. To move an object on the pedigree, simply click and hold your left mouse button on the object, drag the object to its new position, and then release the button. To change the size of an object, place the cursor on the edges of the object until the cursor changes to a double-headed arrow, and then drag the edge as needed to increase or decrease the size of the object.

# **Configuring Pedigree Icons**

Configuring the icons that are used in a pedigree consists at a minimum of the following:

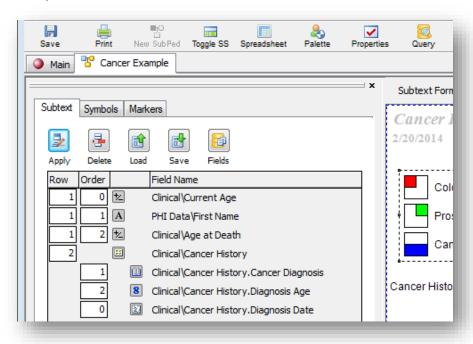
- Configuring the icon subtext, which is the text that is displayed above or below every icon on a pedigree.
- Configuring the icon symbols, which are symbols that graphically represent data about an individual on a pedigree.

Also, if your implementation of Progeny Clinical contains the Marker module, you can also configure the subtext for marker icons, which are the color codes that are used to identify genotypes (marker values) in pedigrees.

### To configure the icons for a pedigree

- 1. On the Progeny main window navigation bar, click the Pedigrees button
- 2. Open the data folder that contains the pedigree for which you are configuring the icons.
- 3. Double-click on the pedigree name to open the Pedigree Drawing window, and on the window's main menu, click View → Icons.
- 4. The Icon window opens in the left pane of the Pedigree Drawing window. The window has at least two tabs—Subtext and Symbols. (By default, the Subtext tab is the active tab.) In addition, if your implementation of Progeny Clinical contains the Marker module, then the window has a third tab—Markers.

Figure 5-10: Icon window, Subtext tab



- 5. Continue to one of the following:
  - a. To configure the icon subtext for a pedigree
  - b. To configure the icon symbols for a pedigree
  - c. To configure icon markers for a pedigree

### To configure the icon subtext for a pedigree

1. Click the Icons button from the top menu then click the Fields button inside the Subtext tab of the Icons window to open the Fields dialog box.

Figure 5-11: Accessing the Fields dialog box from the Subtext configuration menu

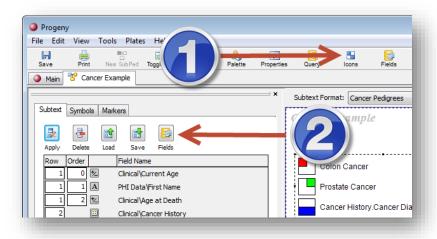
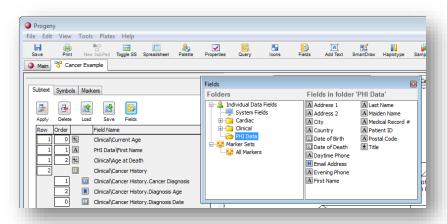
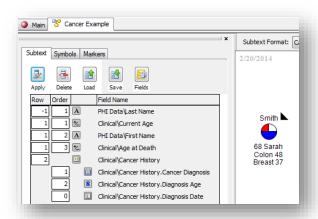


Figure 5-12: Subtext Configuration menu with the Fields dialog box open



- 2. Drag each field that is to be displayed in the icon subtext from the Fields dialog box to the blank Subtext tab. When you are dragging the fields, note the following:
  - The order in which you drag the fields into the subtext configuration menu <u>does not matter</u>. You can
    use the Row column to the left of the field to configure the vertical placement (1 = top row) and the
    Order column to configure the horizontal placement (1 = left-most).
  - You can use negative values in the Row column (ex. -1) to have a subtext row display *above* the pedigree icon.
  - You can display table fields in icon subtext. When you drag a table field from the Field dialog box to the Subtext tab, all the table subfields are included under the table name. To include a table subfield in the icon subtext, enter a number into the Order column for that subfield. If you drag a lookup table to the Subtext tab, then only the lookup table field is displayed on the tab. The lookup table subfields are not displayed.
  - If your data is too long/verbose to display under an icon, you can either turn on the Autosize Subtext based on Data option within the <u>Properties dialog box</u>, <u>Progeny tab</u> or adjust the pedigree's Smart Draw dimensions to allow for the extra horizontal spacing from the <u>Properties dialog box</u>, <u>Dimensions tab</u>.
  - If you add a field in error or decide that you do not want a field incorporated into your subtext, select the field, and click the Delete button.
- 3. After you have ordered the icon subtext, click the Apply button.
- 4. Figure 5-13 below shows the ordering of the fields for icon subtext and the resulting subtext.

Figure 5-13: Example of recording fields for icon subtext on a pedigree



5. When you have finished constructing your subtext format, click the Save button within the subtext configuration window to save this as a subtext format. See <u>Saving and Loading Icon Formats</u>.



Progeny 9 places a high level of importance on saved formats – every configuration (subtext, symbols, datasheets, spreadsheets, etc.) can utilize saved formats to expand the functionality of the Progeny ecosystem.

## To configure the icon symbols for a pedigree

1. Click the Icons button loons from the top menu, click the Symbols tab within the Icons window then click the Fields button inside the Symbols tab to open the Fields dialog box.

Figure 5-14: Accessing the Fields dialog box from the Symbols configuration menu



- 2. Drag each field that is to be represented by a symbol from the Fields dialog box to the blank Symbols tab. When you are adding fields, note the following:
  - You can create symbols for any database field (including <u>System Fields</u>) and/or define a symbol to represent ranges or queries such as individuals with a current age > 50.
  - The order in which you drag the fields is the order from top to bottom in which the symbols are applied to the icon. You may need to experiment a bit with your field orders as when an individual has too many active symbols they can cover each other.
  - If you select a table field, then the "Select Symbol Row Field" dialog box opens. This dialog box lists all the table subfields. You must select the table subfields that are to be used as symbol rows. If you select a lookup table field, then only the lookup field is added to the Symbols tab. The lookup table subfields are not.
  - You can select the same table field multiple times to apply symbols for each individual subfield that is
    within that same table field. For example, if you are creating a pedigree that shows the cancer history
    for a family you could drag the Cancer History field multiple times to apply the symbols for Cancer
    Diagnosis subfields: Cancer Diagnosis = Colon, Cancer Diagnosis = Prostate, and Cancer Diagnosis =
    Breast

Figure 5-14: Select Symbol Row Field dialog box

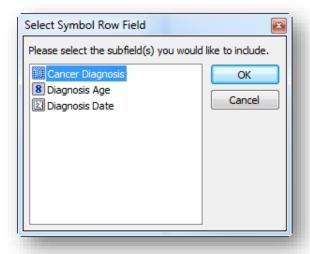
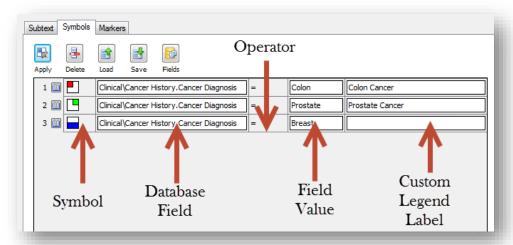
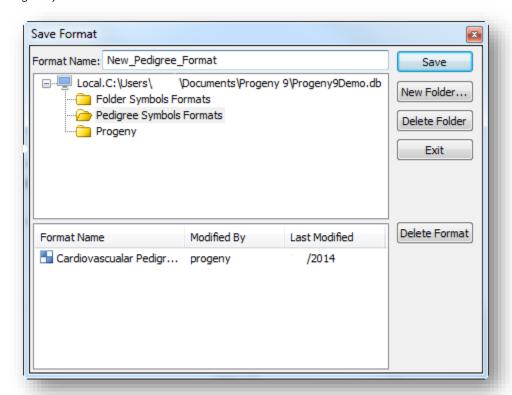


Figure 5-15: Symbols configuration options



- 3. After you have determined which fields you want to display symbols for and have added them to your Symbols configuration menu, do the following for each field (refer to Figure 5-15 for guidance):
  - Click on the left-most column (Symbol) to open a dropdown list from which you can select the symbol to be displayed for each field.
  - Click in the Operator column to open a dropdown list from which you can select the logical operator for the field. (See <u>Query operators</u> for a detailed explanation of the operators.)
  - Enter in the Field Value column the data you want a symbol displayed for.
  - If you have specific requirements for the Symbols Legend or just do not like the default Symbols Legend display schema (ex. Cancer History.Cancer Diagnosis = Breast *OR* Current Age >30) you can use the Custom Legend Label column on the far right to enter a personalized label which will be displayed on the pedigree Symbols Legend instead. Using the two examples in the previous sentence, we could set up our Symbols Legend to instead display easy to understand language for the symbols like <u>Breast Cancer</u> instead of Cancer History.Cancer Diagnosis = Breast or you could configure <u>Patient over 30</u> instead of Current Age > 30.
- 4. If you add a field in error or decide that you want to delete a field, select the field and click the Delete button.
- 5. After you have selected all the necessary icon symbols, click the Apply button
- 6. When you have finished constructing your subtext format, click the Save button within the subtext configuration window to save this as a symbols format. See <u>Saving and Loading Icon Formats</u>.

Figure 5-16: Saving a Symbols Format



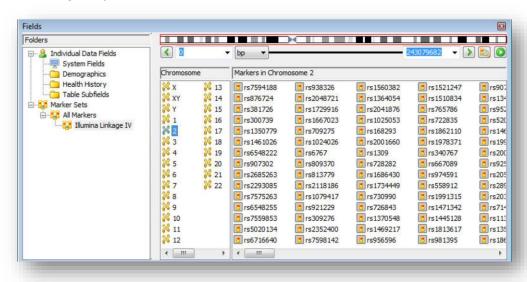


After you configure and save the icon subtext for a pedigree, you can save the subtext as a format, and then load the format for other pedigrees. To see <u>Saving</u> and <u>Loading Icon Formats</u>

### To configure icon markers for a pedigree

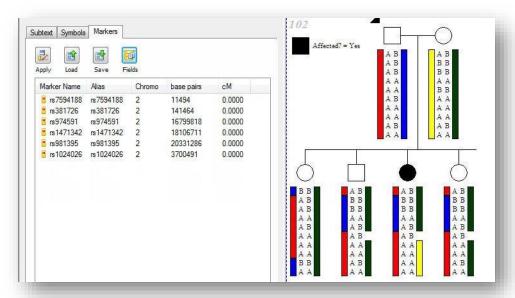
- 1. Open the Markers tab, then click the Fields button to open the Fields dialog box.
  - a. The left pane of the dialog box lists available marker sets in the Progeny database. Select All Markers to open the list of available sets, and then select one of the sets to view the information for the markers contained in that set. The middle pane is the Chromosome List pane, which lists the chromosomes on which you can view or add markers. Select a chromosome to display an image of the chromosome above the top of the right pane. A list of the markers and SNPs that are found in the selected chromosome is displayed below the image in the right pane.

Figure 5-17: Fields dialog box for icon markers



- 2. Drag each marker that is to be displayed on the pedigree from the Fields dialog box to the blank Marker tab. As you select the markers that are to be displayed on the pedigree, note the following:
  - a. Make sure to drag the markers in order of ascending bps. The order in which you drag the markers is the order in which they are displayed under each icon in the pedigree
  - b. If you add a marker in error, or you decide you want to delete a specific marker or all markers, then right-click on the marker name on the Markers tab, and on the context menu that opens, click Remove selected marker. To remove all markers in a singlet, right-click anywhere on the Markers tab, and on the context menu that opens, click Remove all markers.

Figure 5-18: Example of a pedigree showing icon markers



- 3. After you have selected all the necessary icon symbols, click the Apply button.
- 4. If you have any additional icon markers to configure you can do them now otherwise:
  - a. Use the following section as a guide to save your icon markers as a format.
  - b. Click the Save button Save to commit your changes to the pedigree then close out.



After you configure and save the markers format for a pedigree, you can save this as a format, and then load the format for other pedigrees. See <u>Saving and</u>
<u>Loading Icon Formats</u>

# Saving and Loading Icon Formats

After you have configured the necessary icons (subtext, symbols, and markers) for a pedigree, you can save each configuration as an icon format. You can then load this format and apply it when needed to other pedigrees.

#### To save an icon format



The following procedure details how to save an icon subtext format, but by analogy, you can follow this procedure to save an icon symbol format and an icon marker format.

When you have completed your work and are ready to save your subtext format, locate the Save button in the Icons menu. Click the Save button and Progeny will bring up the Save Format window. The first thing you'll see in this window is a list of folders where you can save your subtext format. As seen in Figure 5-20 below, there are three folders created for you by default in Progeny 9. The first two folders are used to automatically manage the formatting throughout the database while the third is provided as a general-purpose folder.

Figure 5-19: Locating the Save Format button within the Icons menu

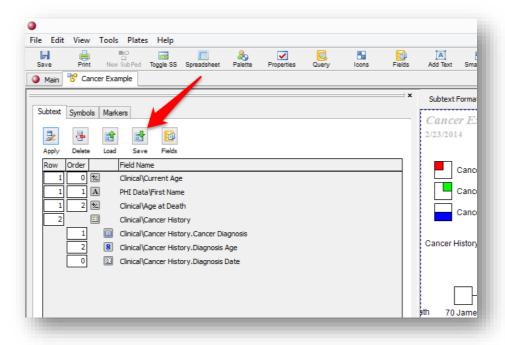
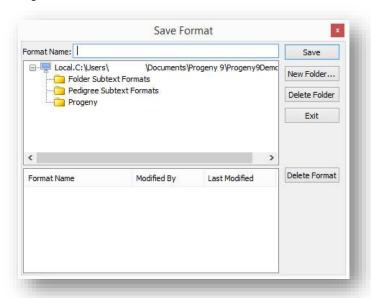


Figure 5-20: Save Format dialog box



Here's how the default folder structure is configured:

- Folder Subtext Formats This is where Progeny saves default formatting for each of your folders if you do not specify a default. These formats will share the name of the folder they represent.
- Pedigree Subtext formats This is a general-purpose folder where formats from imported pedigrees and formats not associated with an object are saved during the Progeny 8 → 9 conversion process.
- Progeny This is the default folder for saving formats.

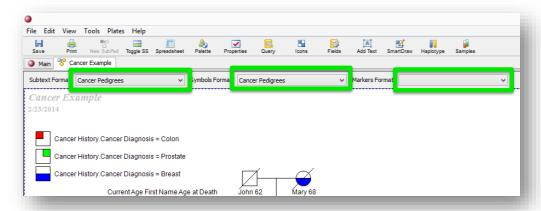
#### To save your format:

- 1. Choose one of the folders above or click the New Folder button to create a new folder.
  - a. Folders can be nested by selecting a folder then clicking the New Folder button.
- 2. Name the format, either by entering a new format name in the top box or selecting an existing format from the bottom box.
- 3. Click the Save button
  - a. If you chose an existing format from the bottom box before clicking Save you will be prompted to overwrite the existing format.

#### To load an icon format

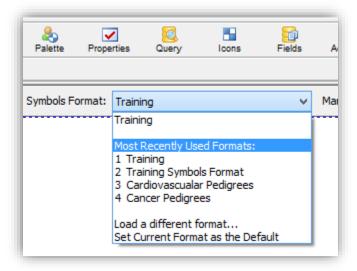
Loading an icon format is an identical process to saving an icon format from the Icons menu, but in Progeny 9 a new dropdown system has be introduced to allow faster access to your saved formats.

Figure 5-21: Icon format dropdowns in the pedigree viewer



In those dropdowns Progeny will display the last five pedigree formats you have used and selecting one will automatically apply that format, while the Load a button, different format button near the bottom, takes you directly into the Load Format menu. Administrators can also use the Set Current Format as the Default to easily make changes to your pedigrees and datasheets.

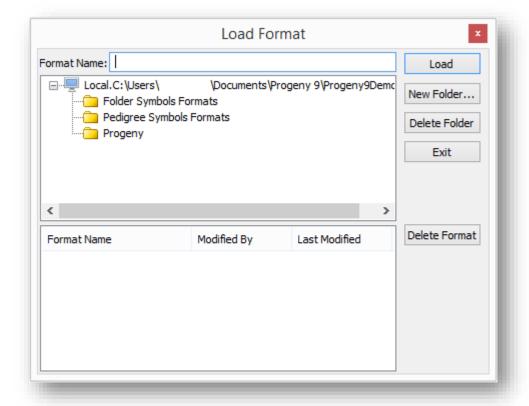
Figure 5-22: Icon format dropdown menu contents



Once you have the Load Format menu open:

- 1. Choose the folder that holds your saved format.
- 2. Select your saved format from the list in the bottom box.
- 3. Click Load. Once complete the format name will appear in its corresponding dropdown menu and will be listed in your most recent formats list.

Figure 5-23: Load Format dialog box



# Manually Drawing a Pedigree

There are four ways to manually draw a pedigree in Progeny:

- Use the palette to add individuals to the pedigree and assign relationships between individuals. See <u>To manually draw a pedigree using the palette</u>
- Use the palette to add your first individual to the pedigree then use the One-click Add function to assign relationships between individuals. See <u>To manually draw a pedigree using One-click Add</u>
- Use the palette to add your first individual to the pedigree then use the Add Relation menu to assign
  relationships between individuals. See <u>To manually drawing a pedigree using the palette and Add Relation
  context menu option</u>
- Create "singlet" individuals outside of a pedigree then move these individuals to a new or existing pedigree. See To manually draw a pedigree after adding and moving individuals to a pedigree

### To manually draw a pedigree using the palette



1. On the Pedigree Drawing window toolbar, click the Palette

button. The Palette dialog box opens.

Figure 5-24: Palette dialog box



- 2. Click the Male or Female icon in the Palette dialog box and then click in the Pedigree Drawing window to add the selected individual.
  - a. This person is also automatically marked as the Proband, but you can change that later from the Individual tab of the Properties menu.



Each time that you click in the Pedigree Drawing window, an icon of the selected gender is added to the pedigree until you either click the Off button in the top-left of the palette or you choose another relation to add to the pedigree.

- 3. Now that your Proband is set go back to the Palette and use the other buttons to fill out the pedigree. Click on a relationship button, then click on an icon that has already been placed on the pedigree.
- 4. When your pedigree is complete, click the Save button to commit your changes.

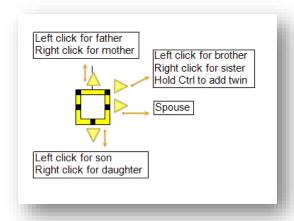
### To manually draw a pedigree using One-click Add



Unless your administrator has set the One-click Add function to be always active, you will need to turn it on from the <u>Progeny tab of the Properties menu</u>.

- 1. Inside a new pedigree either use the Palette from the last section to add an individual or right click the blank pedigree screen and choose Add Individual → {Chosen Gender}
- 2. Click on the individual added in Step 1. The icon will become outlined with a double yellow square and 4 yellow arrows surrounding that 2 arrows pointing right, 1 arrow pointing up and 1 arrow pointing down.

Figure 5-25: Arrows determining relationships to selected individual



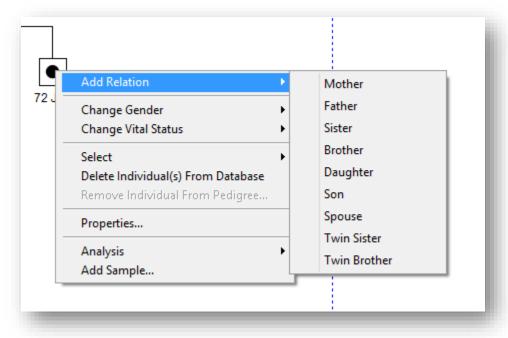
321

- 3. Build your pedigree using the arrows, keeping in mind that left mouse clicks on the arrows will add males and right clicks will add females.
  - a. If you are adding twins and want to specify the specific type (monozygotic or dizygotic), select the steepled line connecting them [the left half of the line usually works better] the choose the twin type from the Line tab of the Properties menu.
- 4. When your pedigree is complete, click the Save button to commit your changes.

### To manually draw a pedigree using the palette and Add Relation menu

- 1. In a new pedigree window open the Palette menu from the <u>previous section</u> and add your first individual. You can now close the Palette.
- 2. Right click on the individual and choose Add Relation (at the top of the list)
  - a. This will open a secondary menu with a list of relations choose one and Progeny will add an individual of the appropriate gender to the pedigree and create the relationship line between them.

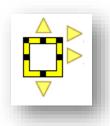
Figure 5-26: The Add Relation context menu options



### To manually draw a pedigree by collecting "singlets"

- 1. After you move individuals to a pedigree (see <u>Moving an Individual to a Pedigree</u>), all of their icons will be piled into the upper left hand corner of the pedigree.
  - a. Sometimes it might look as if you only have a single individual on the pedigree, but all the others are stacked below that single icon.
- 2. Move the icons around using the left mouse button and start laying out the structure of your pedigree.
- 3. When you are ready to start making relationship connectors, left click one of your individuals. You will see the icon become outlined in yellow with black dots around the border.

Figure 5-27: Manual relationship line starting points



- a. Each black dot represents a relationship type:
  - i. Top-Center = Child to Parent
  - ii. Bottom-Center = Parent to Child
  - iii. Top-Corners (both sides) = Sibling to Sibling
  - iv. Middle-Sides = Spouse to Spouse
- 4. Connect your added individuals by clicking and holding on a black dot with the left mouse button.
  - a. The mouse cursor will turn into a set of crosshairs
- 5. Drag the line and over to a second person and place the crosshairs in the center of that person before letting go of the mouse button.
- 6. Once you have your individuals connected with relationship lines you can use the Save button to commit your changes or use any of the methods discussed earlier in this section to add new individuals to the pedigree.
  - a. To manually draw a pedigree using the palette
  - b. To manually draw a pedigree using the One-click Add option
  - c. To manually drawing a pedigree using the Add Relation menu

# Working with Individuals in a Pedigree

For any individual in a pedigree, you can do one or more of the following:

- Change the gender and/or vital status (Alive or Deceased).
- Move an individual or groups of individuals to another location in the pedigree.
- Delete an individual from a pedigree or the database.
- Select an individual based on their familial relationship.
- Correct any Mendelian errors for individuals for which haplotypes are displayed.
- View all the samples that are associated with an individual in a pedigree.

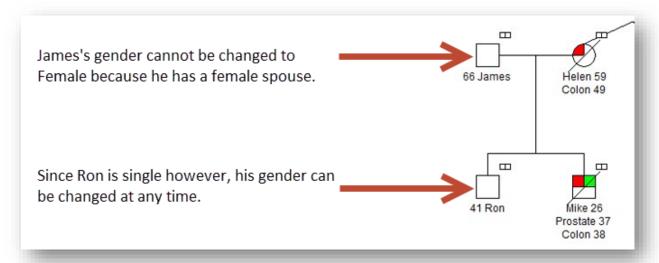
### To change the gender and/or vital status of an individual in a pedigree

You can change the gender of an individual in a pedigree <u>only</u> if the individual is not in a prohibitive relationship. For example, if an individual female in a pedigree has a spouse relationship, you cannot change the individual from female to male.



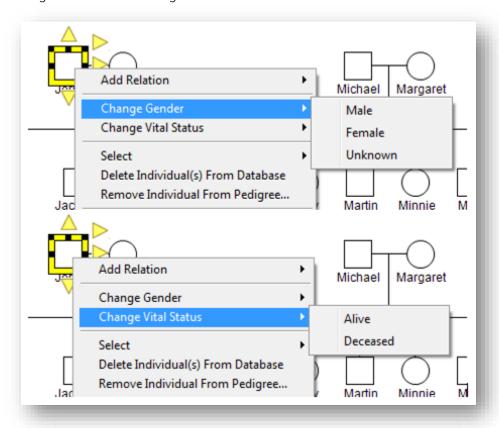
Though currently not possible in Progeny 9, the ability to have same-gender spouse relationships in a pedigree is a feature that is in development and will be coming to Progeny.

Figure 5-28: Changing the gender of an individual in a pedigree



- 1. To change the gender and/or vital status of an individual in the pedigree, select the individual using a left click so that the individual is highlighted with the yellow box.
- 2. Once the individual is actively selected, right click the individual and select Change Gender or Change Vital Status as needed. The icon for the individual will update accordingly.

Figure 5-29: The Change Gender and Change Vital Status context menus:

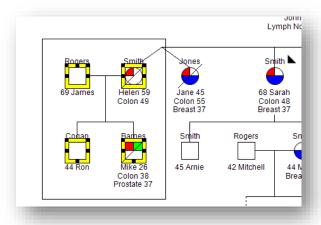


### To move an individual in a pedigree

To move a single individual, simply select the individual with a left click then drag them to a new location in the pedigree. To move a group of individuals, to the following:

- 1. Click and hold the left mouse button on an empty part of the pedigree, and then drag the cursor around the group of individuals that are being moved so that a box is drawn around them.
- 2. Once all the desired individuals are within the "box" release the left mouse button. The "box" will disappear but everyone that was inside it will be highlighted in yellow. See the below image for an example of this.

Figure 5-30: Selecting multiple individuals to move in a pedigree



- 3. To move this group of highlighted individuals, put your mouse icon inside one of the individuals then click the left mouse button and hold it down. Every individual in the highlighted group should still be highlighted.
- 4. While holding the left mouse button down drag the individual you clicked on to your desired new position on the pedigree.



When you are moving individuals in the pedigree, if you reach the edge of the Pedigree Drawing window, the window automatically scrolls, allowing you to move across the entire pedigree without having to drop the individual.

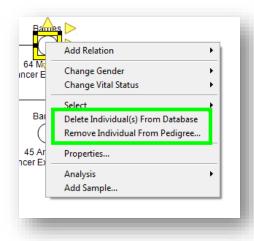
5. When you have reached your destination point for these selected individuals, release the left mouse button and the entire group will settle into their new location. Don't forget to click the Save Save Button to commit your changes.

#### To delete an individual from a pedigree or the database

When removing an individual from a pedigree, you can remove one person at a time or an entire group of individuals. There are also two options for removal – remove from the pedigree or delete from the database.

- 1. Select the individual(s) that will be removed from the pedigree. To do this you can:
  - a. Left click a single individual
  - b. Left click and hold on a blank part of the pedigree then draw a box around a group of individuals
  - c. Left click a single individual then hold down the [CTRL] button on your keyboard and left click any additional individuals you wish to remove.
- 2. To remove the selected individual(s), you can:
  - a. Press the [Delete] key on your keyboard this will delete the selected individual(s) from the database.
  - b. Right click on any of the selected individuals then choose the Delete Individual(s) from Database option.
  - c. If you have only one (1) individual selected right click the icon and an option will be available to Remove Individual from Pedigree. This will extract the individual from the pedigree <u>but keep them in the database</u>. The individual becomes a "singlet" within the database and can be added to another pedigree or left solo.
    - i. This option is not available if more than one (1) individual is selected.
- 3. When you have removed all desired individuals from the pedigree, click the Save button to commit the changes.

Figure 5-31 – Removing an individual from a pedigree or deleting them from the database



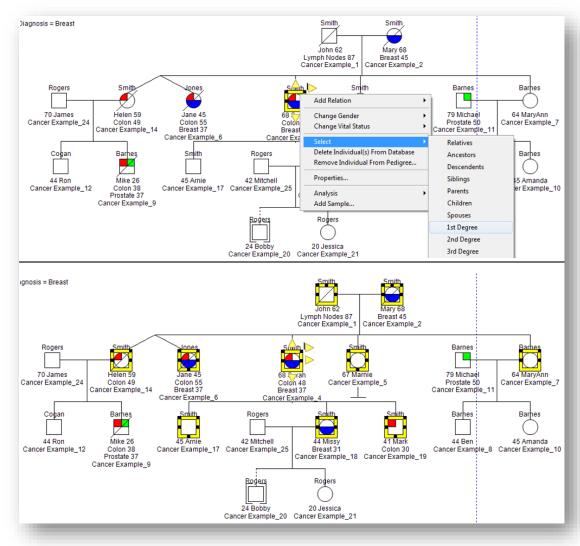
#### To select individuals in a pedigree based on familial relationships

You can select individuals in a pedigree based on their relationships such as first-degree relationships, spouses, and so on. You can also select individuals in a pedigree based on relationships that they do not have, for example, all individuals who are not first-degree relatives for an individual. This is known as a reverse selection.

#### To select an individual based on relationship

Right click the proband then left click the select option in the context menu and choose your desired relationship selection. Progeny will select all individuals in the pedigree that fall into the chosen relationship type. In the figure below, we want to select all first-degree relatives of the chosen individual.

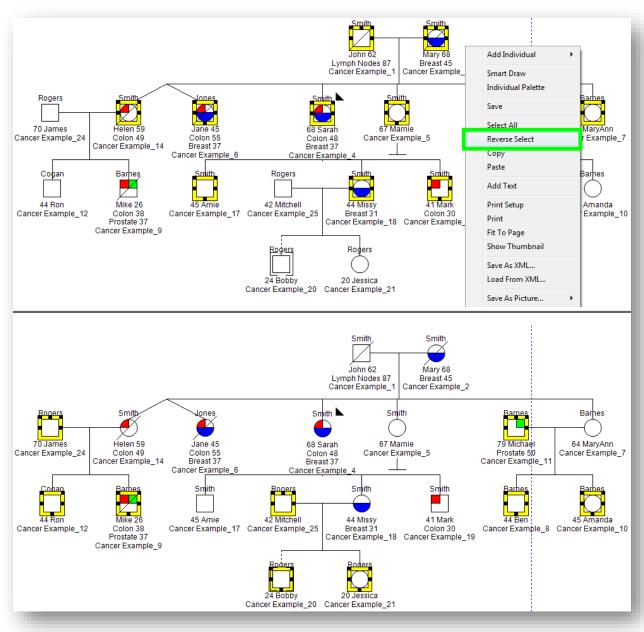
Figure 5-32: Selecting First Degree Individual Relationship



#### To select an individual based on a reverse relationship

To perform a reverse selection, follow the steps in the previous section so that you have a group of individuals selected based on a specific relationship. Next, right click anywhere in the pedigree background to bring up the context menu and choose the Reverse Select option. In terms of a database guery, the individuals now selected are those whose relationship "does not equal" first degree.

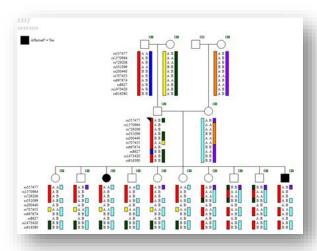
Figure 5-33: Performing a reverse-select when all first-degree relatives of a proband are already selected



#### To correct Mendelian errors for individuals in a pedigree

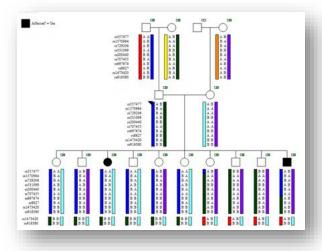
Open the pedigree where you have Mendelian errors in need of correction. Progeny will display these errors as seen in the image below.

Figure 5-34: Pedigree showing Mendelian errors in individuals' haplotypes



Click on the Haplotype Haplotype button along the top of the Progeny window (usually displayed as the second button from the right). Progeny will automatically resolve the Mendelian errors and make the corresponding adjustments on the pedigree. When you are satisfied with the result, make sure to click the Save button to commit your changes.

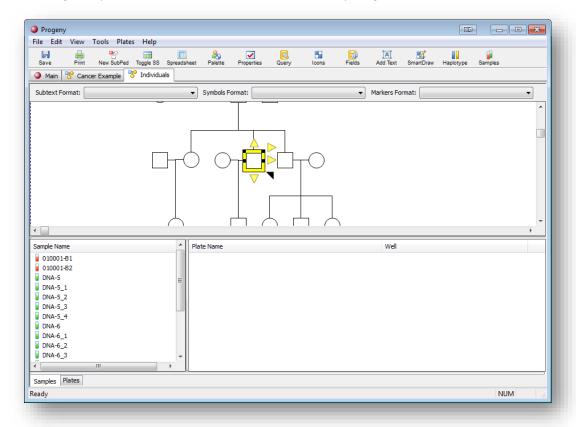
Figure 5-35: Pedigree with Mendelian errors corrected



### To view all the samples associated with an individual in a pedigree

- 1. Select the individual for whom you are viewing the samples.
- 2. Click the Samples button Samples on the Pedigree Drawing window toolbar.
- 3. A Samples Pane and Containers pane opens below the pedigree. The Samples pane lists all the samples that are associated with the selected individual. To view information about a sample, select the sample in the Samples pane.

Figure 5-36: Viewing samples associated with an individual in a pedigree



4. To close the Samples and Containers panes, click the Samples button again.

# Adding an Individual Shadow to a Pedigree

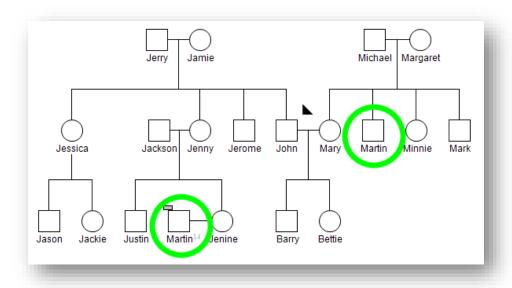
The individual shadow is a feature that indicates a complex marital relationship that otherwise would be shown as a single line that crossed the entire pedigree. For instance, you can add a shadow to display an individual in a second generation who is married to an individual in the first generation. An individual who is a shadow in pedigree is denoted with a thin, dark gray rectangle above its icon. A gray UPN number is displayed to the bottom right of the icon of the shadowed individual, indicating the UPN of the original individual on the pedigree.

Jerry Jamie Michael Margaret

Jessica Jackson Jenny Jerome John Mary Martin Minnie Mark

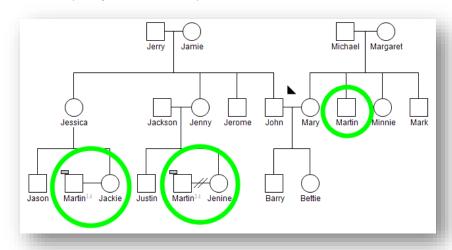
Jason Jackie Justin Jenine Barry Bettie

Figure 5-37: Using an individual shadow to simplify complex pedigrees



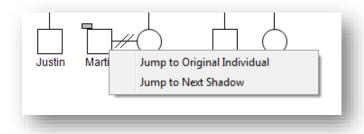
In the example above, Martin (the proband's brother-in-law) marries Jennine (the proband's niece). Now while this will be the source of *years* of family drama it also complicates our perfectly laid out pedigree. Even though the pedigree hooks make it simple to understand the relationships, it doesn't look all that neat. To clean up the pedigree and make it both easier to read and more aesthetically pleasing, we need to replace that relationship line with an individual shadow. This pedigree element acts as a shortcut to and representation of the actual person, displaying the same subtext and symbols yet when the shadow is double-clicked it will open the Individual Data Sheet for its linked person.

Figure 5-38: An Individual in a pedigree with multiple shadows



In larger pedigrees the individual and their shadow might be on opposite ends of the pedigree – to add an additional layer of complexity, an individual can have multiple shadows within a pedigree. With our example family, Martin divorced Jenine then married her cousin Jackie. To represent this on the pedigree, we change the spouse line between Martin and Jenine to display as divorced and create a new shadow next to Jackie. If this was a larger pedigree we could have trouble locating these multiple shadows, so Progeny has built-in controls to make the navigation easier. If you right click on a shadow you will be given an option to Jump to Original Individual, and if there are more than one shadow for the individual you will also have an option to Jump to Next Shadow. By right clicking the original individual, you are given the Jump to Next Shadow option.

Figure 5-39: Navigating between shadows and the original individual



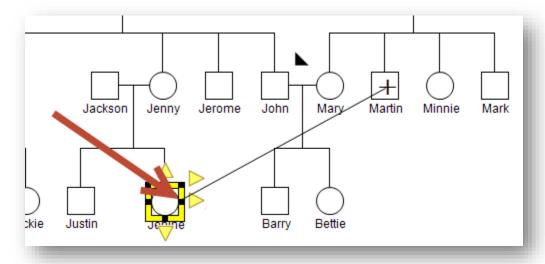


The One-click Add option cannot be selected for a pedigree in which you are creating a shadow individual. To turn off the One-click Add option, open the Progeny tab on the Properties dialog box, and clear the option. See <u>Properties</u> <u>dialog box</u>, <u>Progeny tab</u>.

## To add an individual shadow to a pedigree

- 1. On the pedigree, select one of the spouse icons, press and hold the [CTRL] key, and from either of the spouse handles on the selected icon, drag a line to the other spouse icon. Your mouse icon will become a set of crosshairs.
- 2. Release the [CTRL] key only after the crosshairs are displayed in the center of the spouse icon to which you are dragging the line.
- 3. The shadow of the spouse icon is displayed next to the icon from which you dragged the line. For example, if you drag a line from the spouse John to the spouse Mary, then a shadow of Mary is displayed next to John.

Figure 5-40: Creating a shadow



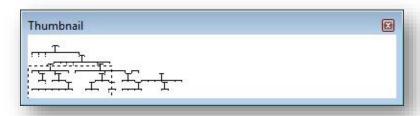
# Creating a Pedigree Thumbnail

You use the Show Thumbnails option to create a pedigree thumbnail, which is a specific area of a pedigree in which to work without having to scroll up or down or left or right.

#### To create a pedigree thumbnail

- 1. On the Pedigree tab of the Properties dialog box, set the needed working scale for the pedigree. For example, set the scale to 200%. See <u>Properties dialog box</u>, <u>Pedigree tab</u>
- 2. Right-click anywhere on a blank section of the pedigree, and on the context menu that opens, click Show Thumbnail. The entire pedigree is displayed in the Thumbnail window.

Figure 5-41: Thumbnail window showing a pedigree



- 3. In the Thumbnail window, click on the area of the pedigree in which you want to work.
  - a. The pedigree is scaled in the Pedigree Drawing window to show only those individuals in the selected area. The Thumbnail window remains open.
- 4. Repeat Step 3 as many times as needed to complete your work on the pedigree.
- 5. After you have completed the necessary work on the pedigree, close the Thumbnail window, and then if needed, on the Pedigree tab of the Properties dialog box, reset the pedigree scale to its original value.

# Chapter 6 Managing Samples

A sample is a part that is representative of a whole or a set of elements that is drawn from and analyzed to estimate the characteristics of a population. A sample can be an unassociated sample, or it can be associated with an individual. Managing samples in Progeny Clinical consists of creating new samples, creating sample datasheets, modifying samples, deleting samples, and importing sample data.

This chapter covers the following topics:

- Manually Adding a New Sample
- **Creating a New Sample Datasheet**
- Modifying a Sample
- **Deleting a Sample**



For detailed information about importing sample data, see Appendix B, Importing Clinical Data.

# Manually Adding a New Sample

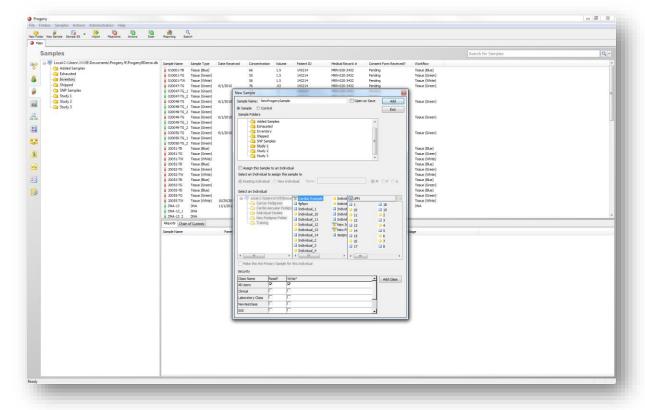
When you add a new sample to a Progeny database, you can add the sample as an unassociated sample, or you can associate the sample with an individual. You can add the sample as a "true" sample (by selecting "Sample" radio button), or you can add the sample as a "control".

## To add a new sample



- 1. On the Progeny main window navigation bar, click the Samples button
- 2. On the Samples window, click the New Samples button New Sample dialog box opens.

Figure 6-1: New Sample window



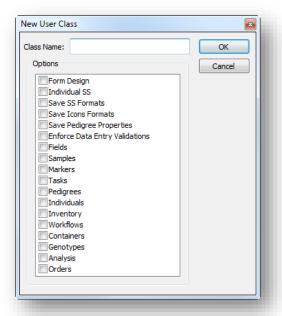
- 3. Enter the name for the new sample and choose the folder where the sample will be added.
- 4. Optionally, select "Open on Save". If you select this option, then after you Add the sample, the sample datasheet opens for the sample.
- 5. Optionally, assign the sample to an individual.
  - a. If you select Existing Individual, then you must select the individual from the available individuals in the database. The individual can be contained in an individual data folder or in a pedigree. If you select a pedigree, then after you select the pedigree, you must select the appropriate individual from the pedigree. In either case, the "UPN" is the default value for identifying the individual, but you can click the Fields button to open the Select Fields dialog box and select another value for identifying the individual (Please reference figure above).
  - b. If you select New Individual, then you must enter a name for the individual, indicate the gender, and select a data folder to which to add the individual.
- 6. If you assigned the sample to an individual, indicate if this sample is to be the primary sample for the individual.



If a sample is marked as the primary sample for an individual, then this is the sample that is exported when selecting an individual for a custom genotype export. See Creating and Running a Custom Genotype Export.

- 7. If you are the Progeny administrator you have the option do one or more of the following security configurations for the individual; otherwise, continue.
  - a. The security privileges for the Sample being created can be set by enabling or disabling the "Read" and "Write" options either for "All Users" within the Progeny database or for the different "user classes" which can be created upon clicking the "Add Class" button. The "Read" and "Write" functions are defined below:
  - b. Read? View the data (name, data in the pedigree datasheet, the pedigree drawing) for the pedigree.
  - c. Write? Write data (change the name, enter data in the pedigree datasheet, draw the pedigree) to the pedigree.
  - d. Click Add Class to open the New User Class dialog box to create a new user class, and then set the pedigree security for the new class.

Figure 6-2: New User Class window



- 8. Give the class a name, check desired options, and then click OK. A message opens indicating the settings for the new class was added successfully. Click OK to close the message and return to the "New Sample" window.
- 9. Click the Add button. A message opens indicating the new sample was added successfully, click OK to close it. If you are assigning this sample to an individual, before clicking "Add", be sure to select this individual in the far-right window pane (Underneath UPN field option).
- 10. When you are finished adding samples, click Exit.

#### To add a new control



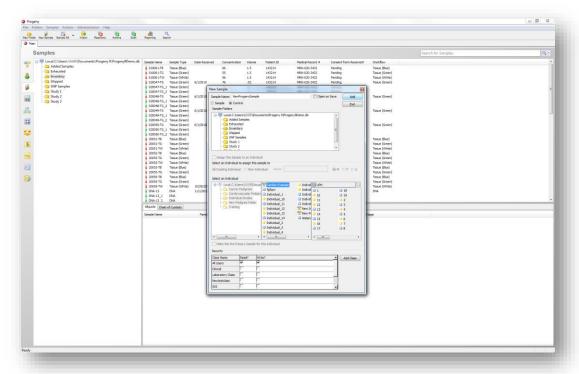
1. On the Progeny main window navigation bar, click the Samples button



2. On the Samples window, click the New Samples button

. The New Sample dialog box opens.

Figure 6-3: New Sample window



- 3. Enter the name for the new sample select "Control."
- 4. Select the Sample folder to which the sample is being added.
- 5. Optionally, Select Open on Save. If you select this option, then after you save the control, the sample datasheet opens for the control.
- 6. Click the Add button. A message opens indicating the settings for the new sample were successfully added, click OK to close the message and return to the Samples window.

## Creating a New Sample Datasheet

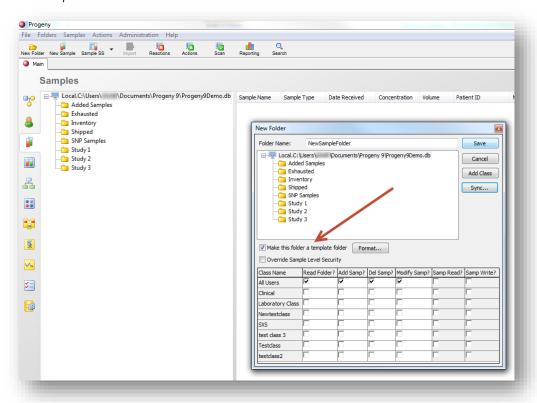
You use a sample datasheet to enter and store data for a sample in a Progeny database. All the samples that are assigned to a sample data folder can use the same datasheet format, or each sample in the folder can use a unique datasheet format. You can create and format a sample datasheet at the time that you add a sample to a data folder, or later.

## To create a sample datasheet



- 1. Create a Sample data folder by clicking the New Folder button.
- 2. Give the folder a name and click Save.
- 3. If all the samples that are assigned to the folder are to use the same sample datasheet format, make sure to select the Make this folder a template folder option; otherwise, clear it. See <u>Adding Sample Data Folders</u>.

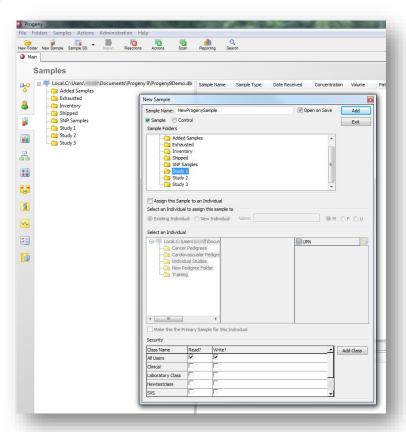
Figure 6-3.1: New Sample Folder window





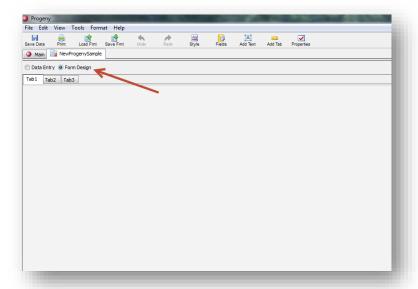
- 4. Create a new sample by clicking the New Sample button. one of the following:
- When you are creating the sample, do
- a. To have a blank datasheet open immediately after you save the sample, select Open on Save. After designating the sample options, click Add to create sample.

Figure 6-3.2: New Sample window



b. Once you are brought to the blank datasheet of Sample, you can click Form Design on this blank datasheet and format the datasheet as needed. If the "Make this folder a template folder" option was selected for this Sample folder, then all the samples that you subsequently add to the folder will use the same datasheet format for entering and storing data; otherwise, you must format a datasheet for each sample that you add to the folder (Please reference figure below).

Figure 6-3.3 Blank Sample datasheet



c. To add all samples to the folder before you create and format the datasheet, do not select "Open on Save" for any sample that you add to the folder. After you have added all the needed samples to the folder, click "Exit", then double-click on any sample in the folder to open a blank datasheet and format the datasheet as needed. If during the creation of this Sample's folder, the "Make this folder a template folder option" was selected, then all samples that are currently added to the folder or that you subsequently add to the folder will use this same datasheet format for entering and storing data; otherwise, you must format a datasheet for each sample in the folder.



See <u>Creating and Formatting Datasheets</u> for detailed information about creating and formatting a sample datasheet.

## Modifying a Sample

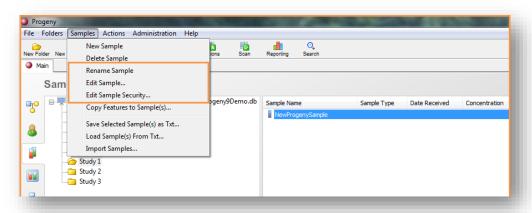
If you are the Progeny administrator, or your administrator has set Write access for the sample in your user account, you can rename a sample and modify the sample (such as assigning a sample to an individual.) As the Progeny administrator, you can also modify the security levels for the sample.

## To modify a sample

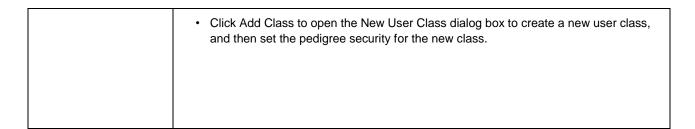


- 1. On the Progeny main window navigation bar, click the Samples button
- 2. Open the sample folder that contains the sample that is being modified.
- 3. Select the sample that is being modified, and on the main menu, click Samples, and then click the appropriate option.

Figure 6-3.3: Sample editing options



Option	Description
Rename sample	Automatically selects the sample name for modification.
Edit Sample	Opens the Edit Sample dialog box in which you can edit the information for the sample (such as assigning the sample to an individual).
Edit Sample Security	<ul> <li>Opens the Sample Security dialog box in which you can do one or both of the following:</li> <li>Modify the security settings for All Users or for the different classes of users.</li> <li>Read? —View the data (name and data in the pedigree datasheet) for the pedigree.</li> <li>Write? —Write data (change the name and enter data to a pedigree datasheet) to the pedigree.</li> </ul>



## **Deleting a Sample**

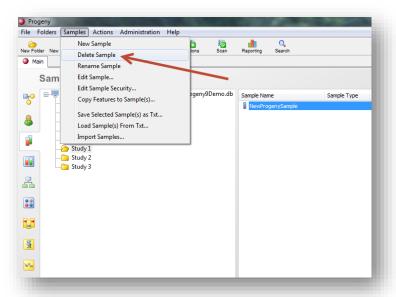
Deleting a sample is a permanent event. After you delete a sample, you cannot recover it from the database.

## To delete a sample



- 1. On the Progeny main window navigation bar, click the Samples button
- 2. Open the sample folder that contains the sample that is being deleted.
- 3. Select the sample that is being deleted (CTRL-click to select multiple samples), and on the main menu, click Samples > Delete Sample(s).

Figure 6-3.4: Deletion of Sample



- 4. A message opens asking you if you are sure that you want to delete the selected samples click Yes.
  - a. The message closes and the selected sample(s) are deleted. Once again you will be returned to the Samples window.

# Section 3 - Progeny Lab

#### This section contains the following chapters:

- Conventions and Sample Management in Progeny Lab
- Managing Markers
- Managing Genotypes
- Linkage Exports
- Family Based Associations
- Hardy Weinberg Test
- Haploview Exports
- Phase Exports
- Custom Genotype Exports
- PLINK Exports
- Helix Tree Exports

# Chapter 1 - Conventions and Samples Management in Progeny Lab

Progeny Lab is software that is designed for the management of whole genome association, targeted genotyping, and linkage studies. Progeny Lab is always sold with Progeny Clinical. As a result, Progeny Lab uses the same terms and conventions as Progeny Clinical. Also, many of the functions that are available in Progeny Lab, such as creating sample data folders, creating samples, importing samples, and so on, have already been detailed in the Progeny Clinical section in this manual, and therefore, do not need to be repeated in detail here. Instead, a summary list of these functions is provided in this chapter, and a link to a detailed discussion about each function is provided.

This chapter covers the following topics:

• Conventions and Sample Management in Progeny Lab

# Conventions and Sample Management in Progeny Lab

Progeny Lab is software that is designed for the management of whole genome association, targeted genotyping and linkage studies. Progeny Lab is always sold with Progeny Clinical. As a result, Progeny Lab uses the same terms and conventions as Progeny Clinical. Also, many of the functions that are available in Progeny Lab, such as creating sample data folders, creating samples, importing samples, and so on, have already been detailed in the Progeny Clinical section in this manual, and therefore, do not need to be repeated in detail here. Instead, a summary list of these functions is provided in this chapter, and a link to a detailed discussion about each function is provided.

- Terms and Conventions Progeny Lab uses conventions in both its terminology and its output to ensure consistency and accuracy when you are manipulating and analyzing genetic data. The terms that are used in Progeny Lab are identical to those used in Progeny Clinical. See <u>Progeny Clinical Terms and Conventions</u>
- Sample Management Like Progeny Clinical, sample management in Progeny Lab consists of adding, modifying, and deleting sample folders, adding, modifying, and deleting samples, copying sample features, creating sample datasheets, and importing sample data. See the following for detailed information in the Progeny Clinical section about each of these sample management functions:
- Adding, modifying and deleting sample folders (including copying samples features) <u>Managing Data</u>
   Folders
- Adding, modifying, and deleting samples (including creating a new sample datasheet) Managing Samples
- Importing sample data Appendix B

## Chapter 2 Managing Markers

Progeny Lab provides functionality for adding markers from an entire genome-wide set (Affymetrix 10K, 100K or 500K, ABI 10cM, SNPlex or TaqMan, Illumina Custom, Golden Gate, 300K, 550K, or 650K, Sequenom, or any other user customized sets), from specific whole chromosomes, or from markers from a specific genomic region. Managing marker sets in Progeny Lab consists of importing marker sets, creating new marker sets and creating marker set formats.

This chapter covers the following topics:

- Markers Window
- Importing Marker Sets
- Saving and Loading an Import Format
- Manually Adding New Markers
- Modifying Marker Information
- Deleting Markers
- Searching for a Marker
- Exporting Marker Sets
- Recalculating Allele Counts for a Marker Set
- Formatting a Marker Datasheet

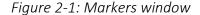
## **Markers Window**

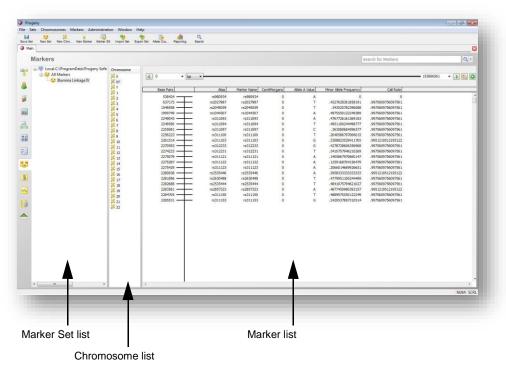
Progeny Lab provides functionality for adding markers from an entire genome-wide set (Affymetrix 10K, 100K or 500K, ABI 10cM, SNPlex or TaqMan, Illumina Custom, Golden Gate, 300K, 550K, or 650K, Sequenom, or any other user customized sets), from specific whole chromosomes, or from markers from a specific genomic region. The Markers window, which displays the information for a loaded marker set, is slightly different in its layout than for the Individuals, Pedigrees, or Samples window.

The left pane displays the Marker Set list, which is a list of marker-sets that are available in a Progeny database. Select All Markers to view all available marker sets. Select one of the marker sets to view information only for the markers in the selected set.

The middle pane displays the Chromosome list, which is a list of the chromosomes in the marker set. Select a chromosome in the list to view an image of the chromosome above the right pane.

The right pane displays the Markers list, which is a list of markers and SNPs for a selected chromosome. From this pane, you can view or update the marker name, the marker alias, and the marker distance information.





## **Importing Marker Sets**

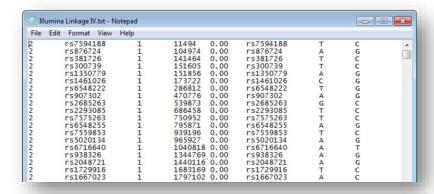
The Import Marker Set function is used to import complete marker sets into a Progeny database. Users can import marker set information to update an existing marker set and allows users to import marker set information to create an entirely new marker set. For a standard import, the import file must be one of three allowed file types—a Standard (Progeny) format, an Illumina OP a Manifest, or an Affymetrix Net Affix Annotation File. (See To import a standard marker set) If the import file is not one of these three allowed file types, then the file must be in a custom format and you must carry out a custom import. (See To import a custom marker set)

## Standard Progeny format

A Standard (Progeny) format must be a text file with a specific structure. Columns 1 through 4 are required. Columns 5 through 8 are optional. Each optional column must be in its allocated column order. To use columns 7 and 8, you must have values in columns 5 and 6.

Column	Description
1	Chromosome – Chromosome on which the microsatellite or SNP is located.
2	Marker Name – Unique alphanumeric name or ID of the microsatellite or SNP.
3	Type – 0 to signify a microsatellite, 1 to signify a SNP
4	Base Pairs – Total number base pairs from the beginning of chromosome at which the marker is located.
5	cM – centimorgan distance from the beginning of chromosome.
6	Alias Name – Alternative display name that is associated with the marker or SNP. For example, Affymetrix genotype calls uses the Affymetrix SNP ID instead of the rs number. If an alias name is not imported, the marker name is displayed in the marker map.
7	Allele A Value – Expected SNP value (ACGT).
8	Allele B Value – Stores expected ACG or T value. Available only for SNPs.

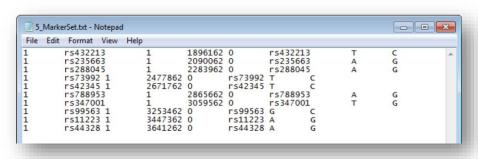
Figure 2-2: Example of a Standard (Progeny) import file



#### Custom import file format

In a custom import, users must identify the columns that contain the data that is to be imported into a Progeny database. The file can contain fields that currently exist in the Progeny database as well as fields that do not currently exist in the Progeny database. The file must contain at least the Marker Name, the Chromo Name and the bp fields. In addition, because all the marker sets that are defined in a custom file can be SNPs, microsatellites, or a mixture of both, the file must also contain a column for designating the marker type (0 indicates a microsatellite, and 1 indicates an SNP).

Figure 2-3: Example of a custom import file

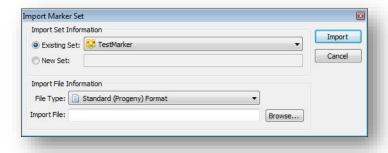


## To import a standard marker set

1. Go to the "Markers" navigation window and from the toolbar, click the "Import Set" button. The "Import Marker Set" dialog box opens.



Figure 2-4: Import Marker Set dialog box

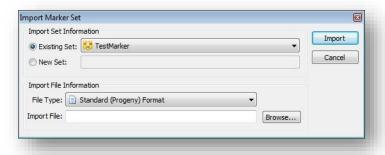


- 2. If you are importing marker set information to update an existing marker set, then select "Existing Set"; otherwise, select "New Set", and then enter a name for the new marker set.
  - a. Select one of the following three file types—Standard (Progeny) format, Illumina OPA Manifest, or Affymetrix Net Affix Annotation File.
- 3. Click "Browse" to browse to and select the marker set import file.
- 4. Click "Import". An "Import Markers" dialog box opens, indicating the progress of the import. When the import process is complete, a "Market Set Complete" message opens, indicating the total number of marker sets that were successfully imported.
- 5. Click "OK" to close the message and return to the "Markers" window.
- 6. The imported marker set is selected in the "Markers" window, and the information for the imported marker set is displayed in the window.

#### To import a custom marker set

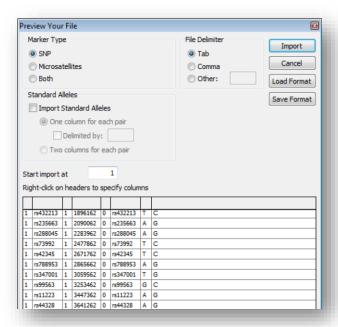
1. Go to the "Markers" navigation window and from the toolbar, click the "Import Set" button Import Set "Import Marker Set" dialog box opens.

Figure 2-5: Import Marker Set dialog box



- 2. If you are importing marker set information to update an existing marker set, then select "Existing Set"; otherwise, select "New Set", and then enter a name for the new marker set.
- 3. Select "Custom Format" from the dropdown "File Type" menu.
- 4. Click "Browse" to browse to and select the marker set import file, and then click "Import".
- 5. An "Import" dialog box opens. The imported file is displayed in the "Import Preview" pane (bottom pane) of the dialog box.

Figure 2-6: Import dialog box



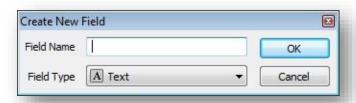
- 6. Select the delimiter that is used for the data in the import file. If you select "Other", you must specify the character that is used for the delimiter.
- 7. Select the marker type that is contained in the import file. If you select "SNP" and your import file contains data for standard alleles, go to Step 7; otherwise, go to Step 8. (HOW DOES THIS MAKE SENSE??)
- 8. Select "Import Standard Alleles", and then indicate how the data is represented.
  - a. One column for each pair The standard alleles are contained within a single column. You must mark Allele AB in the Preview pane.
  - b. Two columns for each pair The standard alleles are in separate columns. You must mark Allele A and Allele B in the Preview pane.
- 9. Specify starting row for the import.
- 10. In the "Import Preview" pane (the bottom pane of the Import dialog box), for each field that exists in the Progeny database, right-click in the column header and manually assign the appropriate heading. When you are finished assigning column headers to the existing fields, if your file contains only fields that are found in the Progeny database, then go to Step 11; otherwise, go to Step 10. (HOW DOES THIS MAKE SENSE??)



The file must contain at least the Marker Name, the Chromo Name, and the bp. If the file contains a mixture of SNPs and microsatellites, then the file must also contain a column for designating the marker type (0 indicates a microsatellite, and 1 indicates an SNP).

- 11. For each field that is not contained in the Progeny database, right-click in the field, and then do one of the following:
  - a. To have the data imported into a new database field, select "Create New Field". The "Add a new Field" dialog box opens. Enter a name for the new field in this dialog box, and optionally, select a different field type. The field will be created and the data for the individual will be imported into this field during the import process.

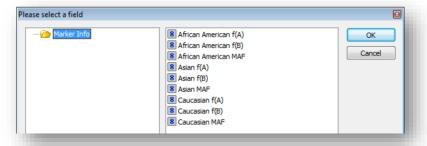
Figure 2-7: Add a New Field dialog box



b. To have the data imported into an existing database field, select "Choose Existing Field". The Select Field dialog box opens, from which you can select an existing marker database field into which the data can be imported.

Figure 2-8: Select Field dialog box

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- c. Additionally, after you label a column, you can do one or both of the following:
  - i. To remove a column from the import, right-click on the column, and on the context menu that opens, select "Ignore This Column".
  - ii. To update a column after it has been labeled, right-click on the column, and on the context menu that opens, select a different column label.
- 12. Click "Import". A "Progeny Import Module" dialog box opens, indicating the status of the import.
- 13. When the import is complete, click "Close".

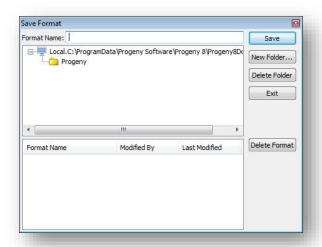
## Saving and Loading an Import Format

After you have set up an import format for a custom marker set, you can save the format. You can then load this saved format and run it on an as-needed basis.

#### To save an import format

- 1. Set up the import format for the custom marker set, but do not click Import. See <u>To import a custom marker</u> set
- 2. Click "Save Format" on the "Import" dialog box.
  - a. The "Save Format" dialog box opens. It provides a default folder named "Progeny" in which to save the import format. (This folder is in the database that you are currently logged in to and you cannot change this location).

Figure 2-9: Save Format dialog box



- 3. Enter a name for the import format, and then do one of the following:
  - a. Select the "Progeny" folder.
  - b. Click "New Folder", and when the "Format Folder" dialog box that opens, enter the following information:
    - Enter the name of the new folder in which to save to format.
    - Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
    - Click Save.

4. Click "Save" in the "Save Format" dialog box. You can then load this saved format and run it on an as-needed basis. See To load an import format

## To load an import format

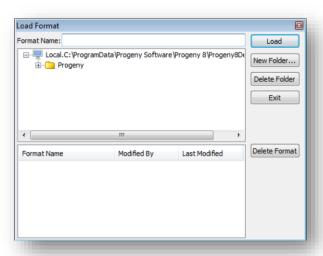
You can a retrieve a saved import format for a custom marker set and load it to import a data file with the appropriate type of data.

1. Open the "Import" dialog box. See To import a custom marker set



2. From the "Import" dialog box, select "Load Format" Load Format" . The "Load Format" dialog box opens.

Figure 2-10: Load Format dialog box



- 3. Open the folder that contains the import format that you are loading, select the format, and then click "Load".
  - a. The "Import" dialog box is refreshed with the loaded format. You can now browse to and select the import file, and then click "Import" to import the data according to the selected format.

## Manually Adding New Markers

You can add custom markers from "scratch" in Progeny Lab by manually entering the information for the markers. The markers can be microsatellites or SNPs. You can add the new markers to an existing chromosome in an existing marker set, or you can add a new marker set and/or new chromosome for the new markers as needed. A new marker set and/or a new chromosome can be added before the new markers are added, or they can be added at the same time that the new markers are being added.



To create a custom marker set by importing a file, see <u>To import a custom marker</u> set.

#### To add a new marker set

When you add a new marker set, you are essentially creating a placeholder for adding markers. All new marker sets are added at the root level (All Markers) for marker sets.

- 1. On the Markers window toolbar, make sure that "All Markers" is selected in the "Marker Set" pane.
- 2. Click the "New Set" button New Set. The "New Set" dialog box opens.

Figure 2-11: New Set dialog box



- 3. Enter a name for the new marker set, and then click Add.
  - a. The "New Set" dialog box closes. The new marker set is displayed in the "Marker List" in the "Markers" window.

#### To add a new chromosome

1. Select the "New Chromosome" New Chro... button from the "Markers" toolbar. The "New Chromosome" dialog box opens.

Figure 2-12: New Chromosome dialog box

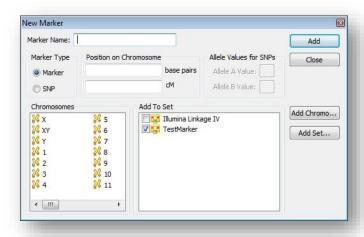


- 2. Enter a name for the new chromosome, and then click "Add".
  - a. The "New Chromosome" dialog box closes. The new chromosome is displayed in the "Chromosome List" in the "Markers" window.

## To manually add new markers

1. Select the "New Marker" New Marker button from the "Markers" toolbar. The "New Marker" dialog box opens.

Figure 2-13: New Marker dialog box



- 2. Enter the name for the new marker.
- 3. Select the appropriate marker type—Marker (microsatellite) or SNP.
- 4. Enter the position for the marker in base pairs for a microsatellite or cM for an SNP.
- 5. If the marker is an SNP, enter the allele values for "Allele A" and "Allele B". (Allowed values are A, C, G, or T).
- 6. Select the chromosome on which the marker is located.



If the chromosome does not exist, then click "Add Chromo" to open the "New Chromosome" dialog box and add a new chromosome.

- 7. Do one of the following:
  - a. To add the marker at the "All Markers" level, do not select any marker sets in the "Add to Set" pane.
  - b. To add the marker to a specific marker sets, select these marker sets in the "Add to Set" pane.



If the marker set does not exist, then click "Add Set" to open the "New Set" dialog box, and add a new marker set.

8. Click "Add". The "Add a New Marker" dialog box closes. The new marker is added to the marker list on the selected chromosome.

# **Modifying Marker Information**

You can modify the following information for a marker in a marker set:

- The base pair size
- The marker name (Available only at the "All Markers" level)
- The marker alias (Applicable only for custom or imported markers.)
- The cM distance

### To modify marker information

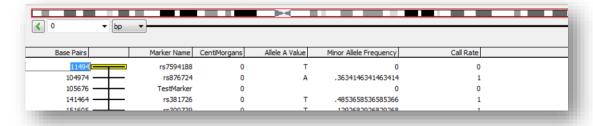
1. Locate the marker for which you are modifying the information in the "Markers" window.



If you are modifying the marker name, then you must locate the marker at the "All Markers" level. You can modify the marker alias only for custom or imported markers.

2. Click once on the information that you modifying to select it, and then modify the information as needed.

Figure 2-14: Example of a marker bps selected for modifying



# **Deleting Markers**

You can delete a marker from a marker set, or you can delete a marker from a Progeny database. When you delete a marker from a set, the marker is not deleted from the database. The marker is still listed at the "All Markers" level, and can be added back to the set, or to another set. To delete a marker from the database, you must delete the marker at the "All Markers" level.

#### To delete a marker from a set

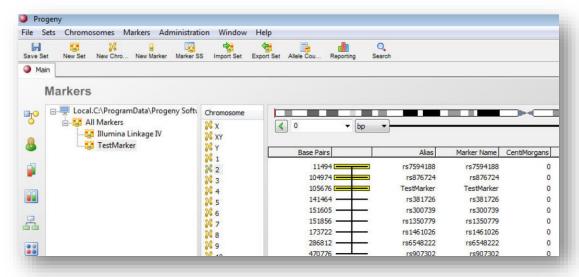
1. Locate the markers that you are deleting in the "Markers" window.



The markers must be in a marker set, and not at the "All Markers" level.

- 2. Select the marker that is to be deleted (CTRL-click to select multiple markers), and then do one of the following:
  - a. Press the [Delete] key on your keyboard.
  - b. Right-click on any of the selected markers, and on the context menu that opens, click "Remove Selected Marker(s)" from the Set.

Figure 2-15: Example of a markers selected for deletion from a set



- 3. A message opens asking you if you are sure that you want to delete the selected markers.
- 4. Click "Yes". The message closes and the markers are deleted.

#### To delete a marker from the database

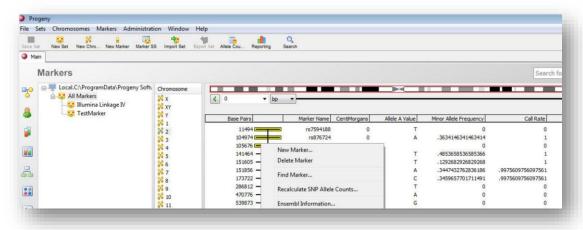
- 1. Make sure that you are logged in to the database from which you are deleting the marker.
- 2. Locate the marker that you are deleting in the "Markers" window.



The markers must be at the "All Markers" level.

- 3. Select the marker that is to be deleted (CTRL-click to select multiple markers), and then do one of the following:
  - a. Press the [Delete] key on your keyboard.
  - b. Right-click on any of the selected markers, and on the context menu that opens, click "Delete Marker".

Figure 2-16: Example of a markers selected for deletion from the database



- 4. A message opens asking you if you are sure that you want to delete the selected markers.
- 5. Click "Yes". The message closes and the markers are deleted.

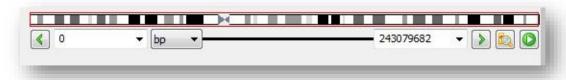
## Searching for a Marker

Two methods are available to you for searching for a specific marker in a Progeny database—the "Marker Filter" and the "Find Marker" function.

### To search using the Marker Filter

The "Marker Filter" is in the right pane of the "Markers" window above the "Markers" list. A figure of a chromosome is displayed above the filter. When the "Markers" window first opens, the entire chromosome is surrounded by a red location rectangle. The bp range for all the markers in the "All Markers" list is displayed.

Figure 2-17: Marker Filter



#### 1. Do one of the following:

- a. On the dropdown list, select the option by which to search (bp, cM, name, or alias information), enter the search criteria, and then click the "Run" button to search for the markers that meet the criteria.
- b. Carry out a contextual search for the marker or markers. See Contextual Search.

The markers that meet the search criteria are displayed in the "Markers" list, and the location rectangle is moved to the appropriate area on the chromosome. If you entered a distance as a search criterion (either bp or CM), then you can click the Jump buttons at either end of the filter to continue searching based on the distance criteria. For example, if your search filter is set from 1000000 bp to 2000000 bp, then click the "Jump Forward" button to change the filter to 200000 through 3000000 bp and update the Markers list accordingly.

## To search using the Find Marker function

1. Click the "Find Marker" button below the chromosome image. The "Find Marker" dialog box opens.

Figure 2-18: Find Marker dialog box



2. Enter the name or alias of the marker for which you are searching.



You must enter the marker name or alias exactly as it is stored in the database, but the search is not case-sensitive. For example, RS238976 and rs238976 return the same marker.

3. Select either one of "Current Set" or "Entire Database".



Any marker set can be selected if you select "Entire Database". If you select "Current Set", make sure that the set that you want to search is indeed the selected set in the "Markers" window.

4. Click "Find". If the marker is found, Progeny switches the view to the corresponding set and chromosome and highlights the marker.

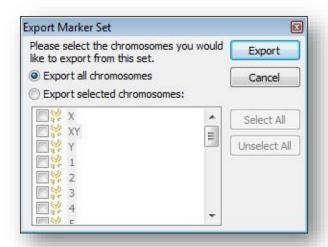
## **Exporting Marker Sets**

You can export marker sets for chromosomes including positioning information. You can export marker sets for all chromosomes, or for only selected chromosomes. A "Marker Set" export file is formatted according to the "Standard (Progeny) file format".

### To export marker sets

- 1. Select the marker set from the "Markers" window for which you are carrying out the export.
- 2. On the "Markers" window toolbar, click the "Export Set" button Export Set. The "Export Marker" Set dialog box opens.

Figure 2-19: Export Marker Set dialog box



- 3. Select "Export all chromosomes", or select "Export selected chromosomes", and then select the chromosomes for which the marker sets are to be exported.
- 4. Click "Export". The "Save As" dialog box opens. The "Save as Type" field is automatically populated with the file type of text (.txt) and you cannot change this value.
- 5. Browse to the location in which the exported file is to be saved and enter a name for the file in the "File Name" field.
- 6. Click "Save". A message opens indicating the export operation was successful.
- 7. Click "OK" to close the message and return to the "Markers" window.

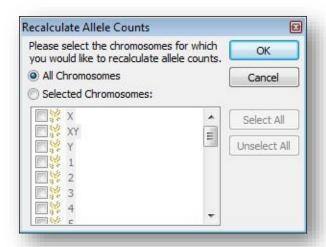
# Recalculating Allele Counts for a Marker Set

The Allele Count function is used to update the counts of AA, AB, BB, and NoCall counts for each SNP, which, in turn, allows for the calculation of the Hardy Weinberg p-value, the minor allele frequency, the degree of heterozygosity, and call rates. If genotypes have been manually updated users can also recalculate allele counts.

#### To recalculate allele counts for a marker set

- 1. Select the marker set for which you are recalculating the allele count from the "Markers" window.
- 2. Click the Export Set Allele Cou... button from the "Markers" window toolbar. The "Recalculate Allele Counts" dialog box opens.

Figure 2-20: Recalculate Allele Counts dialog box



- 3. Select "All chromosomes", or select "Selected chromosomes", and then select the chromosomes for which the allele counts are to be recalculated.
- 4. Click "OK". A "Recalculate Allele Counts" dialog box opens, indicating the progress of recalculating the allele counts.
- 5. When the recalculation is complete, click "Close" to close the dialog box and return to the "Markers" window.

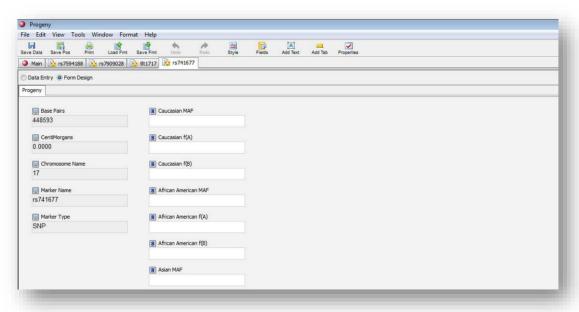
# Formatting a Marker Datasheet

Marker datasheets can be used to enter and store data for a marker in a Progeny database. When you create a new marker, a marker datasheet is automatically created for the marker. All marker datasheets are created with the same five system fields and the same nine Marker Info fields. You can add more system fields and Marker Info fields to a marker datasheet. All markers that are in the same set use the same datasheet format.

#### To format a marker datasheet

- 1. Select the marker set for which you are formatting the datasheet from the "Markers" window.
- 2. Double-click one of the markers in the marker set. The marker datasheet opens.

Figure 2-21: Marker datasheet



3. If needed, you can add more fields to the datasheet. See <u>Datasheet Basics</u>.

# Chapter 3 Managing Genotypes

Progeny Lab provides functionality for importing genotypes directly from supported file formats such as Illumina, Affymetrix, ABI, Sequenom, as well as other formats. You can also manually enter genotypes.

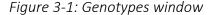
This chapter covers the following topics:

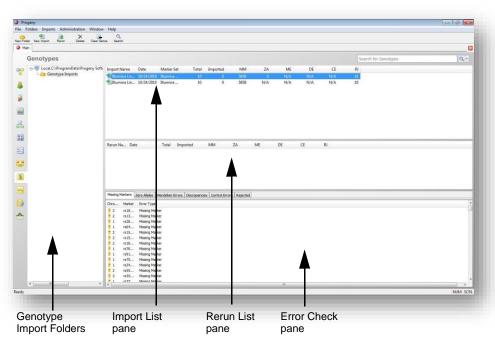
- The Genotypes Window
- Importing Genotypes
- Error Checks for Genotype Imports
- Rerunning a Genotypes Import
- Manually Entering Genotype Data
- Clearing Genotype Data

## The Genotypes Window

The "Genotypes" window displays the information for imported genotypes and differs slightly in its layout in comparison to the "Individuals", "Pedigrees", and "Samples" windows.

- The left pane displays the genotype import folders, which are the folders that hold all the genotypes that have been imported into a Progeny database.
- The upper right pane displays the Genotype Import list, which is a list of all the imports that are contained in a specific genotype import folder. The pane displays the following information (from left to right) for an import—the import name, the import date, the name of the marker set used for comparison and error checking the import, the total number of markers in the set, the total number of markers imported, the total number of missing markers (MM) in the import, the total number of zero alleles (ZA) in the import, the total number of Mendelian errors (ME) in the import, the total number of discrepancies in the import, the total number of control errors (CE) in the import, and the total number of data lines rejected from the import.
- The middle right pane displays the "Rerun Import" list, which is a list of all the imports that were rerun for an original import. The pane displays the same information for a rerun as the top pane displays for an original import.
- The bottom right pane, or "Error Check" pane, displays expanded information for each error check that was carried out for an original import or a rerun. To view the results of a specific type of error check for an import or rerun, select the import or rerun in the appropriate pane of the "Genotypes" window, and then open the relevant tab in the "Error Check" pane. (By default, the "Missing Markers" tab is the active tab for the pane when the Genotypes window first opens.)





# **Importing Genotypes**

Progeny Lab provides functionality for importing genotype files in one of three ways:

- Using a Standard Import format.
- Using one of the following third-party formats—Illumina, ABI, or Affymetrix.
- Using a Custom Import format.

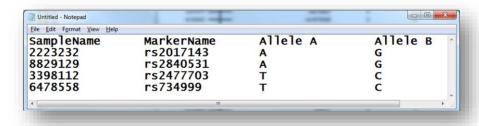
The Custom Import Format is the preferred option because you can specify the exact layout of the file you are importing, which includes designating the data that is contained in each column of the file to ensure compatibility.

### Standard Import format

A Standard Import format must be a tab-delimited text file with a specific structure.

Column	Description
1	The sample name. All alphanumeric characters are allowed, and there is no limit on the number of characters.
2	The marker name. All alphanumeric characters are allowed, and there is no limit on the number of characters.
3	The value for allele 1.  Note: For microsatellites, there are no restrictions on the data that can be displayed in this column. For SNPs, the value can be either an A or a B, or it can be an A, C, G, or T.
4	The value for allele 2.  Note: For microsatellites, there are no restrictions on the data that can be displayed in this column. For SNPs, the value can be either an A or a B, or it can be an A, C, G, or T.

Figure 3-2: Example of a Standard import file



### Illumina Final Report Format

The "Illumina Final Report Format" is an output format that is generated by the Illumina platform. Because of the number of iterations that can cause compatibility issues with the Progeny application, Progeny strongly recommends that the "Custom Import Format" is used instead. By using the "Custom Import Format", users can specify the file layout and import the data as indicated in the file.

#### ABI

The ABI format is an output format that is generated by the "Applied Biosystems by Life Technologies" platform. Because of the number of iterations that can cause compatibility issues with the Progeny application, Progeny strongly recommends that the "Custom Import Format" is used instead. By using the "Custom Import Format", users can specify the file layout and import the data as indicated in the file.

### Affymetrix files

The "Affymetrix CHP file", the "Affymetrix GDAS Text Output Format", and the "Affymetrix GTYPE Text Output Format" are output formats that are generated by the "Affymetrix" platform.

- Each "Affymetrix CHP" file contains a single sample. The "Affymetrix" file name is either the exact name of the sample name or it can also contain an underscore with additional text, such as '223339\_axt3343'. If the file contains an underscore with additional text, Progeny ignores this additional text during the import. For instance, '223339\_axt3343' is imported as '223339'.
- Because of the number of iterations that can cause compatibility issues with the Progeny application,
  Progeny strongly recommends that the "Custom Import Format" in lieu of either the "Affymetrix GDAS Text
  Output Format" or the "Affymetrix GTYPE Text Output Format" is used. By using the "Custom Import
  Format", users can specify the file layout and import the data as indicated in the file.

### Custom import file format

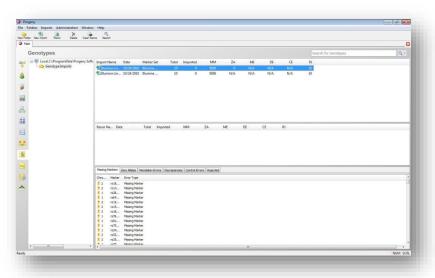
A custom genotype file can have one of three formats:

- One row per call Calls in row, with the following columns: Unique ID, Marker Name, Allele 1, and Allele 2.
- One row per sample Samples in rows with markers in columns
- One row per marker Markers in rows with samples in columns

### To import genotypes

1. On the navigation bar, click the Genotypes button to open the Genotypes window.

Figure 3-3: Genotypes window



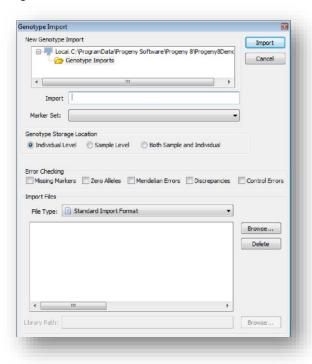
2. Select the folder in which to store the imported genotypes.



You can add a new folder if needed. See <u>Adding, Editing and Deleting Database</u> <u>Folders</u>.

3. On the Genotypes window toolbar, click the New Import button. The Genotype Import dialog box opens.

Figure 3-4: Genotype Import dialog box



- 4. Define the import.
  - a. In the top pane of the dialog box, select the folder in which to import the genotype data.
  - b. In the "Import" field, enter a name for the genotype import.
  - c. From the "Marker Set" dropdown list, select the marker set against which the genotype data is to be verified.
  - d. Progeny verifies the imported genotype data by matching the markers that are contained in the import against the markers that are listed in the selected marker set. All error checking that is carried out on the import is based on this selected marker set. For example, markers that are defined in the marker set, but are not found in the import file, are flagged as missing markers.



This step assumes that you have previously created the necessary marker set for comparison. If you need to create or edit a marker set, see <u>Managing Markers</u>.

5. Indicate the level at which the genotype information is to be stored – with the individual only, with the sample only, or at both the individual and sample level.



If you select at both the individual and sample level, you are duplicating the genotype data in the database. Therefore, sample level only would be the preferable option.

a. Select the type of error checking that is to be carried out—Missing Markers, Zero Alleles, Mendelian Errors, Discrepancies, and/or Control Errors.



For a complete explanation of each type of error checking method, see <u>Importing</u> <u>Genotypes</u>.

- 6. Continue to one of the following:
  - a. To complete a standard import.
  - b. To complete a custom import.

### To complete a standard import

1. Select one of the following standard import formats - Standard Import Format, Illumina Final Report Format, Affymetrix CHP File, Affymetrix GDAS Text Output Format, or Affymetrix GTYPE Text Output Format.



If you are using an Affymetrix Library, then you must also specify the path for the library, In the Library path field, click "Browse" to open the "Browse for Folder" dialog box and browse to and select the folder for the library.

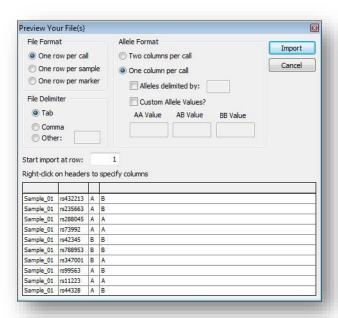
- 2. Click "Browse" to browse to and select the genotype import files.
- 3. Click "Import". A dialog box opens, indicating the progress of the import. When the process is complete, a message opens indicating that the import was successfully completed.
- 4. Click "OK" to close the message and the dialog box. The user will then be returned to the "Genotypes" window. The import is listed in the top pane of the "Genotypes" window.
- 5. To view the results of a specific type of error check for the import, select the import in the top pane of the "Genotypes" window, and then open the appropriate tab in the "Error Check" (bottom) pane.

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### To complete a custom import

- 1. Select "Custom Format" for the file type.
- 2. Click "Browse" to browse to and select the genotypes import file. An Import dialog box opens. The imported file is displayed in the "Import Preview" pane (bottom pane) of the dialog box.

Figure 3-5: Import dialog box



- 3. Indicate the format of your custom file.
- 4. Select the delimiter that is used for the data in the import file. If you select "Other", you must specify the character that is used for the delimiter.
- 5. Indicate the format for the allele calls in the file.
  - a. Two columns per call Each allele call is in a separate column. Continue to Step 7.
  - b. One column per call Both allele calls are in the same column. Continue to Step 6.
- 6. Optionally, do one or both of the following:
  - a. Select "Alleles delimited by", and then specify the delimiter.
  - b. Select "Custom Allele Values", and then enter the appropriate allele values.
- 7. Specify the starting row for the import.

- 8. In the "Import Preview" pane (the bottom pane of the Import dialog box), for each field, right-click in the column header and manually assign the appropriate heading.
  - a. One row per call requires a "Sample Name" column and a "Marker Name" column. If "Two columns per call" is selected, you must indicate the "Allele A" and "Allele B" columns. If "One Column per call" is selected, you must indicate the "Allele AB" column.
  - b. One row per sample requires a "Sample Name" column. If "Two columns per call" is selected, you must indicate the "Allele A" and "Allele B" columns. If "One Column per call" is selected, you must indicate the "Allele AB" column.
  - c. One row per marker requires a "Marker Name". If "Two columns per call" is selected, you must indicate the "Allele A" and "Allele B" columns. If "One Column per call" is selected, you must indicate the "Allele AB" column.
- 9. Click "Import". A dialog box opens, indicating the progress of the import. When the process is complete, a message opens indicating that the import was successfully completed.
- 10. Click "OK" to close the message and the dialog box. The user is returned to the "Genotypes" window. The import is listed in the top pane of the "Genotypes" window.
- 11. To view the results of a specific type of error check for the import, select the import in the top pane of the "Genotypes" window, and then open the appropriate tab in the "Error Check" (bottom) pane.

# **Error Checks for Genotype Imports**

Progeny Lab provides functionality for importing genotypes directly from supported file formats such as Illumina, Affymetrix, ABI, Sequenom, as well as other formats. When you import genotypes, an option is available for carrying out specific error checks - Missing Markers, Zero Alleles, Mendelian errors, Discrepancies, Control Errors, and Rejected - on the data. "Error Check" results are displayed in the bottom pane of the "Genotypes" window. To view the results of a specific type of error check for an import, select the import in the top pane of the "Genotypes" window, and then open the appropriate tab in the "Error Check" (bottom) pane. (By default, the "Missing Markers" tab is the active tab for the pane.)

Fig. Foldow: Imports: Administration: Window Help

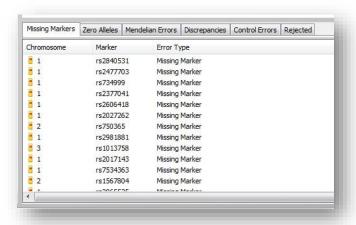
Fig. Foldow: Imports: Administration: Imports: I

Figure 3-6: Selected genotype import and error checking results displayed in bottom pane

### Missing Markers tab

When you import genotypes, you must select a marker set to compare the import file against. In a "Missing Markers" error check, Progeny compares the markers that are contained in the import file against the markers that are listed in the selected marker set. All error checking that is carried out on the import is based on this selected marker set. For example, markers that are defined in the marker set, but are not found in the import file, are flagged as missing markers.

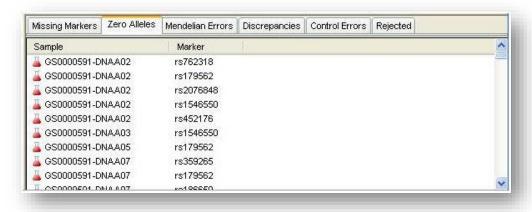
Figure 3-7: Missing Markers tab



#### Zero Alleles tab

In a "Zero Alleles" error check, Progeny checks each sample for all markers and identifies any markers that contain zero alleles, which occurs when the value for a microsatellite cannot be determined. As a result, a zero (0) is displayed for the marker value. The "Zero Alleles" tab displays the sample name and the corresponding marker that contains a zero allele.

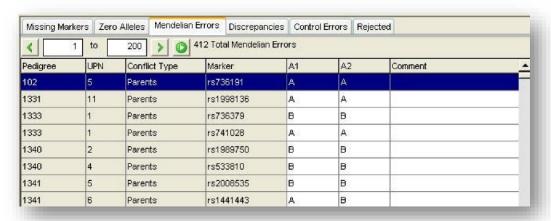
Figure 3-8: Zero Alleles tab



#### Mendelian Errors tab

In a "Mendelian Errors" check, Progeny compares the data for each allele against the relationship structure of the pedigree and verifies the compatibility. Any discrepancies are identified as "Mendelian Errors", and are displayed in the "Mendelian Errors" tab.

Figure 3-9: Mendelian Errors tab



The first four columns in this tab (Pedigree, UPN, Conflict Type, and Marker) are system-generated fields and therefore, are not editable. The final three columns (A1, A2, and Comment) are editable fields. You can change the value of either "Allele 1" or "Allele 2" to resolve any conflicts as well as enter comments in the "Comment" field for record keeping purposes. You can edit a Mendelian error directly from the "Mendelian Errors" tab, or you can edit the Mendelian mirror directly in the pedigree.

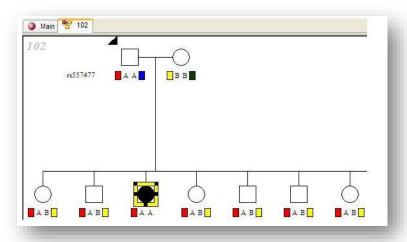
#### To edit a Mendelian error from the Mendelian errors tab

- 1. Select the error that you are correcting from the "Mendelian Errors" tab.
- 2. Edit the value as needed, and then right-click on the edited value, and on the context menu that opens, click "Save Changes".
  - a. Repeat Step 1 and Step 2 to correct all the Mendelian errors.
- 3. Select a row for which you corrected the Mendelian error (CTRL-click to select multiple rows), right-click on any of the selected rows, and when the context menu opens, click "Rerun Mendelian Checks on All Selected Rows". The Mendelian checks are rerun for all the rows to ensure that the error was properly corrected.
- 4. A message opens, indicating that the Mendelian error rerun was successfully completed. It also indicates the number of errors that were corrected, and that the corrected rows have been disabled. Click "OK" to close the message and return to the "Mendelian Errors" tab.
- 5. Optionally, to delete a disabled row, select the row (CTRL-click to select multiple rows), right-click on any selected row, and from the context menu that opens, select "Delete All Selected Rows".

#### To edit a Mendelian error in the pedigree

- 1. From the "Mendelian Errors" tab, right-click on the error that you are editing, and from the context menu that opens, select "Open Error to Pedigree".
  - a. The pedigree that contains the individual with the Mendelian error opens. The specific marker is loaded in the pedigree, and the allele information is displayed as a haplotype for the affected individual.

Figure 3-10: Opened pedigree with Mendelian error

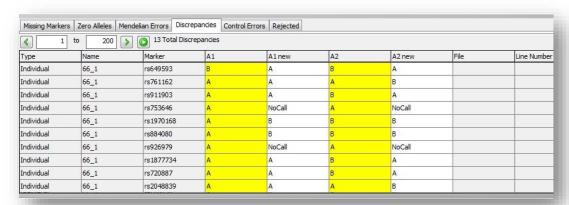


- 2. To edit the value, click on the allele value and enter the correct value.
- 3. Close the pedigree, making sure to answer the prompt about saving the changes. After correcting the error in the pedigree, the user is returned to the "Mendelian Errors" tab. The row that contained the error is disabled.
- 4. Right-click on any corrected error row, and from the context menu that opens, click "Rerun Mendelian Checks on All Selected Rows". The Mendelian checks are rerun for all the row to ensure that the error was properly corrected.
  - a. A message opens, indicating that the Mendelian error rerun was successfully completed. It also indicates the number of errors that were corrected, and that the corrected rows have been disabled.
- 5. Click "OK" to close the message and to return to the "Mendelian Errors" tab.
- 6. Optionally, to delete a disabled row, select the row (CTRL-click to select multiple rows), right-click on any selected row, and from the context menu that opens, click "Delete All Selected Rows".

### Discrepancies tab

The "Discrepancies" tab lists any discrepancies that were found between an original genotype import and a rerun of the import. The first three fields on the tab (Pedigree, UPN, and Marker) are system fields, and are not editable. The next four fields display the original value for allele 1 (A1), the new value for A1, the original value for allele 2 (A2), and the new value for A2, respectively. When the tab first opens, the original values for A1 and A2 (highlighted in yellow on the tab) are the values that are stored in the database. You can resolve these discrepancies from the "Discrepancies" tab, or you can resolve these discrepancies from the pedigree. You can also export the information that is displayed in the "Discrepancies" tab to a text file.

Figure 3-11: Discrepancies tab





To change the view on the tab, enter new values for the beginning and ending discrepancy range, and then click the "Run" button. You can click the "Jump" buttons at either end of the filter to update the display according to the range that you specified. For example, if your search filter is set from 1 to 200 to show

the first 200 discrepancies, then click the "Jump Forward" button to change the range to 200 to 400 and update the display accordingly.

#### To resolve discrepancies from the Discrepancies tab

- 1. To resolve the discrepancy for an allele, click on the new value for the allele.
  - a. The new value for the allele is now highlighted in yellow, indicating this is the value that is to be stored for the allele in the database, and the original value now has no background color (white), indicating that the original value is to be rejected.
- 2. After you have determined which values to store in the database and which values to reject for both A1 and A2 for each marker, select a row (CTRL-click to select multiple rows), and then right-click on any selected row, and from the context menu which opens, select "Resolve Selected Discrepancies".
  - a. The selected discrepancies are removed from the "Discrepancies" tab and the changes that you made are saved in the Progeny database. The selections that you made are used when data is exported for analysis.

#### To resolve discrepancies from the pedigree

- 1. Right-click on the discrepancy in the "Discrepancies" tab, and from the context menu which opens, select "Open Pedigree to Error".
- 2. The pedigree is displayed onscreen.
- 3. Make the changes directly in the displayed pedigree.
- 4. Close the pedigree to return to the "Discrepancies" tab.
- 5. Select the changed value in either the "A1 New" or "A2 New" column.
  - a. The new value for the allele is now highlighted in yellow, indicating this is the value that is to be stored for the allele in the database, and the original value now has no background color (white), indicating that the original value is to be rejected.
- 6. After you have you have determined which values to store in the database and which values to reject for both A1 and A2 for each marker, select a row (CTRL-click to select multiple rows), and then right-click on any selected row, and from the context menu which opens, select "Resolve Selected Discrepancies".
  - a. The selected discrepancies are removed from the "Discrepancies" tab and the changes that you made are saved in the Progeny database. The selections that you made are used when data is exported for analysis.

#### To export discrepancies to a text file

- 1. Right-click on any row in the "Discrepancies" tab, and from the context menu that opens, select "Export to File".
- 2. Specify the name and location for the export file. The exported file contains all the discrepancies, including the original call and the change that was committed to the database.

#### Control Errors tab

The "Control Errors" tab lists any controls where the expected value for A1 and A2 is different than the value which was returned in the genotype file that was imported. The expected calls are listed in the columns "A1 Control" and "A2 Control". The returned incorrect calls are listed in A1 and A2 columns. To export the control errors to a text file, right-click anywhere on the tab, and from the context menu that opens, click "Export to File". After a control is identified as an error, all the data for the marker in the genotype import will be rejected. The resulting rejected markers are listed on the "Rejected" tab with the reason "Entire Marker invalid due to control error."

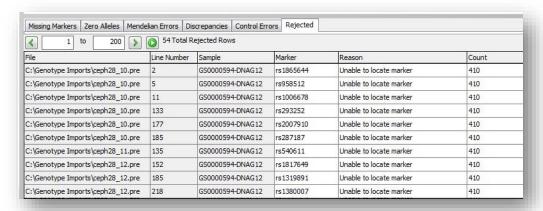
### Rejected tab

The "Rejected" tab lists all the data in the genotype import file that was rejected for one of the following reasons:

- There is no match in the database with a sample name that appears in the genotype import. The sample name must exist in the database to store the genotype information.
- There is no match in the database with a marker that appears in the genotype import. The marker must exist in the database to store the genotype information.
- If duplicate markers and samples are contained in the genotype import, and the values for both are zero, the second marker/sample entry is rejected.
- If the expected Allele A value or Allele B value is not stored for a given SNP or marker in the "Markers" module, then the SNP or marker is rejected.
- If a field cannot be updated during the import process (for example, another user is updating a "Sample Name" field during your import process), then the data that references the field is rejected.
- If any database error occurs during the import process, then the field that failed to record data is listed in the "Rejected" tab along with the value that could not be recorded.

None of the fields in this tab are editable. The first three columns (File, Line, and Sample) list the file that contained the rejected data, the line number where the error check occurred in the file, and the sample that contained the error, respectively. The remaining three columns (Marker, Reason, and Count) list the marker that contains the error, the reason for the rejection, and the count for multiple error rows, respectively.

Figure 3-12: Rejected tab



To change the view on the tab, enter new values for the beginning and ending rejected range, and then click the "Run" button at either end of the filter to continue searching based on the range that you specified. For example, if your search filter is set from 1 to 200 to show the first 200 rejected samples, then click the "Jump Forward" button to change the range to 200 to 400 and update the display accordingly.

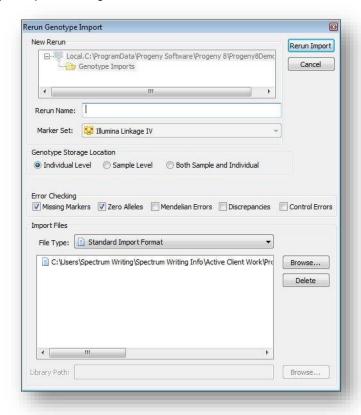
# Rerunning a Genotypes Import

After you run an original genotype import, and then make all the needed corrections to the import, you can run the import again.

### To rerun a genotype import

1. From the "Genotypes" window, in the "Import List" pane, select the original import that is to be rerun, and then from the window toolbar, select the "Rerun" button. The "Rerun Genotype Import" dialog box opens.

Figure 3-13: Rerun Genotype Import dialog box



- 2. When this dialog box opens:
  - a. The marker set that was used for the original import is automatically selected for the rerun, and you cannot change this value.
  - b. The "Error Checking" options that were selected for the original import are also automatically selected, however, you can change these selections.
  - c. The file type that was used for the original import is automatically selected for the rerun, however, you can change this selection.



If you are using an Affymetrix Library, then you must also specify the path for the library, In the Library path field, click "Browse" to open the "Browse for Folder" dialog box and browse to and select the folder for the library.

- 3. The files that were used for the original import are displayed in the lower pane of the dialog box. The list of files can be changed if needed, including deleting files, and selecting new files.
- 4. After you have made all the necessary modifications for the rerun, click "Rerun Import".
- 5. A dialog box opens, indicating the progress of the import. When the process is complete, a message opens indicating that the import was successfully completed.
  - a. Click "OK" to close the message and the dialog box.
- 6. You return to the "Genotypes" window. The import is listed in the middle pane of the "Genotypes" window. Any discrepancies between the original import and the rerun are listed in the "Discrepancies" tab.

# Manually Entering Genotype Data

Instead of importing genotype data into a Progeny database, you can manually enter data.

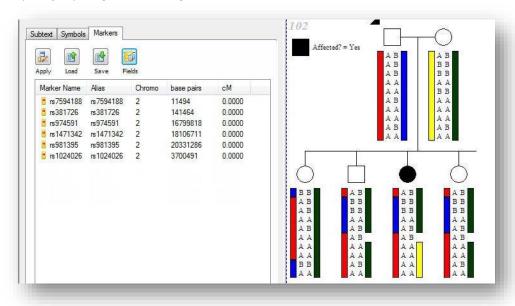


If you manually enter genotype data, error checking of the data will not automatically be carried out.

Four options are available for manually entering genotype data:

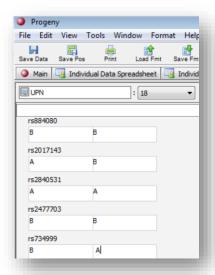
1. You can enter genotype data using icon markers on a pedigree. See <u>To configure icon markers for a pedigree</u>.

Figure 3-14: Example of a pedigree showing icon markers



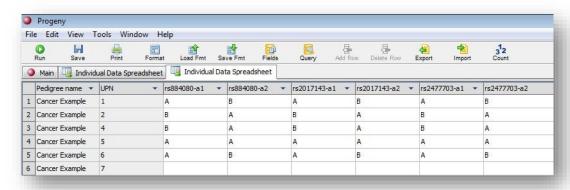
2. You can enter genotype data into the marker fields on a datasheet for an individual. See <u>Creating and</u> Formatting Datasheets.

Figure 3-15: Marker data in an individual datasheet



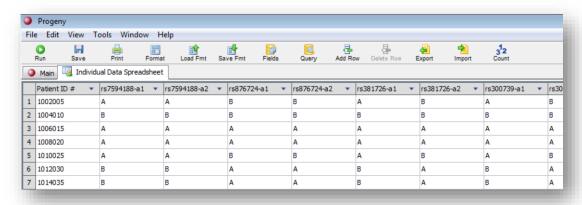
3. You can enter genotype data directly into the marker fields in a spreadsheet for an individual. For example, select the pedigree from the "Pedigrees" window, and then from the toolbar, select the "Individual Spreadsheet" button. Select the system fields of "Pedigree Name" and "UPN" and the appropriate marker fields, and then run the spreadsheet. The marker field is automatically split into two fields representing the paternal allele in a1 and the maternal allele in a2. See Generating a Spreadsheet.

Figure 3-16: Entering genotype data into an individual spreadsheet



4. You can import data from a text file into the marker fields in a spreadsheet for an individual. For example, select a field that matches the individuals in the spreadsheet to the individuals in the text file such as "Patient ID #", and the appropriate marker fields, and then run the spreadsheet. The marker field is automatically split into two fields representing the paternal allele in a1 and the maternal allele in a2. After running the spreadsheet, import the marker data. See To import data into a spreadsheet.

Figure 3-17: Importing genotype data into an individual spreadsheet



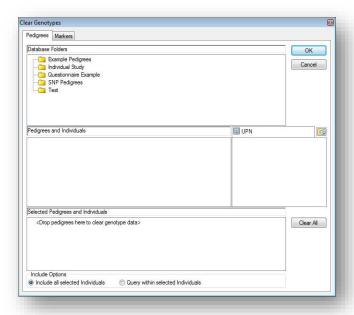
# Clearing Genotype Data

The "Clear Genotypes" function is used to clear genotype data from selected individuals in selected pedigrees.

### To clear genotype data

1. On the Genotypes window toolbar, click the Clear Genotypes Clear Genos button. The Clear Genotype dialog box opens. By default, the Pedigrees tab is the active tab.

Figure 3-18: Clear Genotypes dialog box, Pedigrees tab



- 2. In the Database Folders (top) pane of the dialog box, select the folder that contains the pedigrees for which the genotype data is to be cleared.
- 3. In the Pedigrees and Individuals (left middle) pane of the dialog, select the pedigree (CTRL-click to select multiple pedigrees) for which the genotype data is to be cleared.
  - a. The individuals that are contained in a selected pedigree are displayed in the right middle pane of the dialog box. By default, the UPN is the identifier used for the individuals.



Optionally, you can click the Fields button next to the Individuals list to open the Select Field dialog box and select a different identifier, such as the Global ID, for individuals in a pedigree.

- 4. Drag the selected pedigrees to the Selected Pedigrees and Individuals (bottom) pane of the dialog box.
- 5. Do one of the following:
  - a. To delete the genotype data for all individuals in all selected pedigrees, select include all selected individuals.
  - b. To delete the genotype data only for those individuals who meet selected criteria, select Query within selected individuals, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.



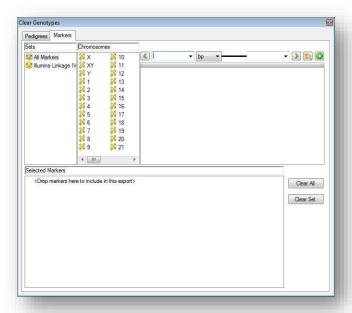
For detailed information about defining a query, see <u>Database Queries and</u> Searches.

- 6. Open the Markers tab, and then do one of the following:
  - a. To clear an entire marker set, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
  - b. To clear the all the markers for a specific chromosome, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
  - c. To clear only specific markers, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See <u>Searching for a Marker</u>.

Figure 3-19: Clear Genotypes dialog box, Markers tab



- 7. Open the Pedigrees tab, and then click OK on the tab.
  - a. A message opens, asking you if you are sure that you want to clear the genotype data.
- 8. Click Yes to close the message.
  - a. A dialog box opens, indicating the status of clearing the genotype data.
- 9. When the status is complete, click Close to close the dialog box and return to the Genotypes window.

# Chapter 4 Linkage Exports

Progeny Lab provides all the functionality needed for managing genetic linkage studies, including managing linkage exports. Managing linkage formats consists of creating and running linkage exports, saving linkage export formats for running on an as-needed basis, rerunning linkage exports, importing LOD scores, and deleting linkage exports.

This chapter covers the following topics:

- Creating and Running a Linkage Export
- Saving and Loading a Linkage Export Format
- Viewing and Rerunning a Linkage Export
- Importing LOD Scores
- Displaying LOD Scores on a Pedigree
- Deleting a Linkage Export

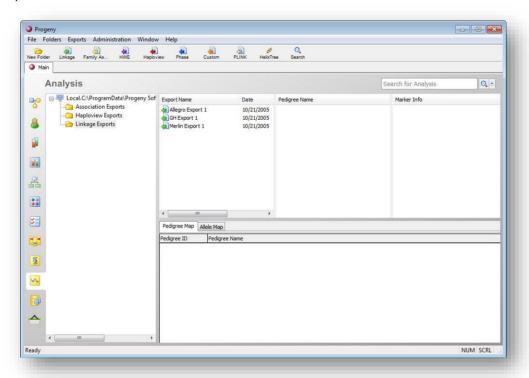
## Creating and Running a Linkage Export

You can export MLINK, Gene hunter, MEGA2, Allegro, and Merlin linkage formats in Progeny Lab. You must first create the linkage export, and then you can run the linkage export.

### To create a linkage export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 4-1: Analysis window



2. On the Analysis window, select the folder in which to the linkage export is to be saved.

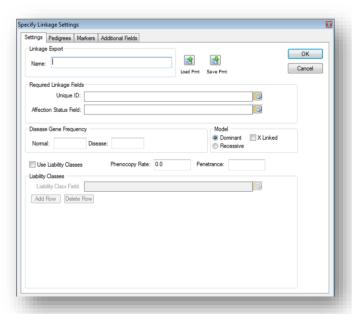


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the Linkage button Linkage. The Specify Linkage Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary linkage settings on all four tabs of the dialog box. See:
  - a. Specify Linkage Settings dialog box, Settings tab
  - b. Specify Linkage Settings dialog box, Pedigrees tab
  - c. Specify Linkage Settings dialog box, Markers tab
  - d. Specify Linkage Settings dialog box, Additional Fields tab
- 5. After you specify the necessary linkage settings on all four tabs, return to the Settings tab, and then click OK on the tab.
- 6. Continue To run a linkage export

## Specify Linkage Settings dialog box, Settings tab

Figure 4-2: Specify Linkage Settings dialog box, Settings tab



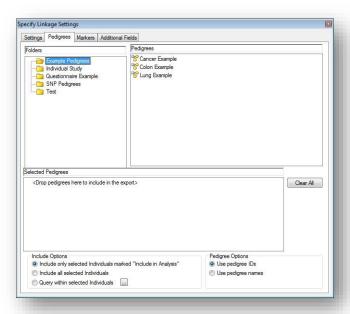
1. Enter the settings for the linkage export.

Option	Description	
Linkage Export		
Linkage Export Name	Name or description of the linkage export.	
Required Linkage Fields		
Unique ID	Click the Fields button to the right of the Unique ID field to open the Select Field dialog box and select the field that represents the unique ID for everyone that is to be included in the linkage export.	
Affection Status Field	Click the Fields button to the right of the Affection Status Field to open the Select Field dialog box and select the field that represents the affected status for everyone that is to be included in the linkage export.	
Disease Gene Frequency		
Normal Disease	Enter the appropriate values for Normal and Disease gene frequency.  Note: The gene frequencies, when totaled, must equal 1.0.	
Model		
Dominant/Recessive	Select one value.	
X Linked	Select if relevant.	
Use Liability Classes	Select if applicable, and then click the Fields button to the right of the Liability Class Field to open the Select Field dialog box and select the field that holds the liability class values. A table opens in the Liability Classes (bottom) pane of the dialog box for identifying the values for the liability classes. To add a row to the table, click Add Row.  Note: For X-Linked models, specify three female values and two male values; otherwise, specify three values and leave the remaining blank.	
Phenocopy Rate Penetrance	Available only if Use Liability Classes is not selected.	

2. Continue specifying any other linkage export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a linkage export.

#### Specify Linkage Settings dialog box, Pedigrees tab

Figure 4-3: Specify Linkage Settings dialog box, Pedigrees tab



- 1. In the Folders (top left) pane of the dialog box, select the folder that contains the pedigrees for which the linkage export is being created.
- 2. In the Pedigrees (top right) pane of the dialog, select the pedigree (CTRL-click to select multiple pedigrees) for which the linkage export is being created.
- 3. Drag the selected pedigrees to the Selected Pedigrees (bottom) pane of the dialog box.
- 4. Select one of the following:
  - a. To include only those individuals in a pedigree who have been specifically marked for inclusion in an analysis, select Include only selected individuals marked "Include in Analysis." See <u>To select</u> individuals in a pedigree for inclusion in an analysis
  - b. To include all individuals in all the selected pedigrees are included in the linkage export, select include all selected individuals.
  - c. To include only those individuals in the export linkage who meet specific criteria, select Query within selected individuals, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.



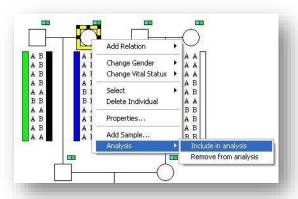
#### For detailed information about defining a query, see Database Queries and **Searches**

- 5. Select one of the following:
  - a. Use Pedigree IDs—Converts a text-based pedigree name into a numerical ID.
  - b. Use pedigree names—Use pedigree names as-is.
- 6. Continue specifying any other linkage export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a linkage export.

## To select individuals in a pedigree for inclusion in an analysis

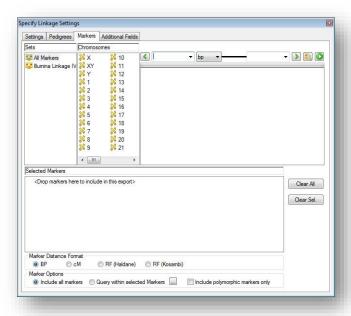
If you select the option Include only selected individuals marked "Include in Analysis," then you must open each affected pedigree, and manually select the appropriate individuals. You can select a single individual, or CTRL-click to select multiple individuals, and then right click on any selected individual, and on the context menu that opens, click Include in analysis.

Figure 4-4: Selecting individuals for analysis



#### Specify Linkage Settings dialog box, Markers tab

Figure 4-5: Specify Linkage Settings dialog box, Markers tab



#### 1. Do one of the following:

- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



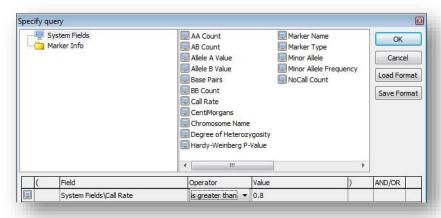
To search for a specific marker or markers, you can use the marker search functions. See Searching for a Marker.

- 2. Specify the intermarker distance format that is to be exported.
  - a. BP or cM exports the exact distance between the markers in base pairs or centimorgans, respectively.
  - b. RF (Haldane) and RF (Kosambi) exports the distance between the markers based on the Haldane recombination factor or the Kosambi recombination factor, respectively.
- 3. Select one of the following:
  - a. To include all the markers in the Selected Markers pane in the linkage export, select include all markers.
  - b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.



For detailed information about defining a query, see <u>Database Queries and Searches</u>. If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

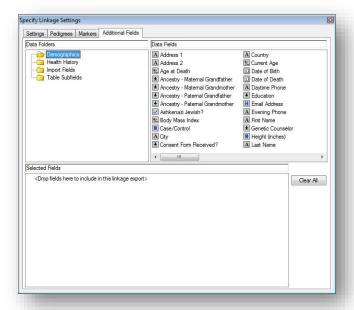
Figure 4-6: Additional marker filtering



- 4. Optionally, select Include polymorphic markers only to further filter the selected markers and export only those markers that have at least one heterozygous genotype.
- 5. Continue specifying any other linkage export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a linkage export.

#### Specify Linkage Settings dialog box, Additional Fields tab

Figure 4-7: Specify Linkage Settings dialog box, Additional Fields tab

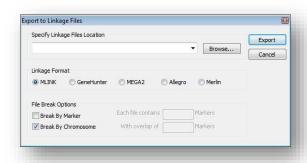


- 1. Specify additional individual level database fields that are to be included in the linkage export.
- 2. Continue specifying any other linkage export settings as needed; otherwise, return to the Settings tab, click OK and continue below.

## To run a linkage export

After you specify the necessary linkage settings on all four tabs, you must return to the Settings tab, and then click OK on the tab. The Export to Linkage Files dialog box opens.

Figure 4-8: Export to Linkage Files dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the linkage export files are be stored.



If you have previously run linkage exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

- 2. Select the appropriate linkage format.
- 3. Select one or both file break options.
  - a. Break by Marker Indicate how many markers are to be contained in each linkage file. If you select this option, you must also indicate the number of overlapping markers in each file.
  - b. Break by Chromosome Creates a new file for each chromosome that is exported.
- 4. Click Export. A dialog box opens, indicating the status of the linkage export.
- 5. When the export is complete, click Close to close the dialog box and return to the Analysis window.
- 6. Load the files that were created by the linkage export into your analysis software to produce LOD files that you can import into Progeny. See <a href="Importing LOD Scores">Importing LOD Scores</a>.

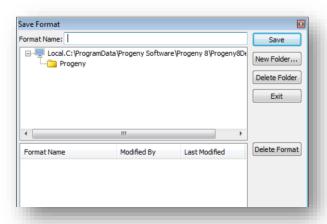
# Saving and Loading a Linkage Export Format

After you have set up a linkage export format, you can save the format. You can then load this saved format and run it on as-needed basis.

## To save a linkage export format

- 1. Set up the linkage export format but do not run the format See Creating and Running a Linkage Export.
- 2. Click the Save Format Save Fmt button on the Specify Linkage Settings dialog box.
  - a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 4-9: Save Format dialog box



- 1. Enter a name for the linkage export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
- 2. Click Save, then click Save on the Save Format dialog box.
- 3. You can then run this saved format on an as-needed basis. See To load a linkage export format.

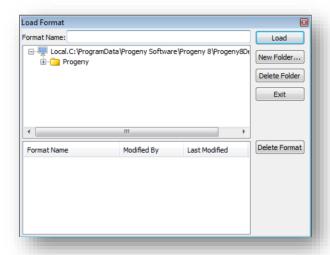
## To load a linkage export format

1. Open the Specify Linkage Settings dialog box. To create a linkage export, steps 1-3.



2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 4-10: Load Format dialog box



- 3. Open the folder that contains the linkage export format that you are loading, select the format, and then click Load.
  - a. The Specify Linkage Export Settings dialog box is refreshed with the loaded format. You can modify the format as needed (for example, modify the information on the Pedigrees tab), and export the linkage format.

# Viewing and Rerunning a Linkage Export

You can view information about any linkage exports that were previously run on the Analysis window, and you can rerun any linkage export as necessary.

### To view and rerun a linkage export

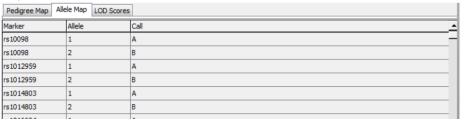
- 1. On the Analysis window, select the linkage export that you want to view.
  - a. Three tabs open in the bottom pane of the window—Pedigree Map, Allele Map, and LOD Scores. By default, the Pedigree Map tab is the active tab.
- 2. Open and view the information on each tab as necessary.
  - a. The Pedigree Map tab displays the mapping of the original pedigree name to the pedigree ID generated for the export file. These values are not editable; however, you can sort this information by clicking the heading for either the Pedigree ID or the Pedigree Name.

Figure 4-11: Pedigree Map tab



b. The Allele Map tab displays the mapping of the original calls for each marker to the allele number generated for the export file. These values are not editable; however, you can sort the data by clicking the heading for either the Maker, Allele, or Call.

Figure 4-12: Allele Map tab



- c. You can also right-click on either of these tabs and open a context menu with options for copying the information on the tab, exporting the information on the options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.
  - i. The LOD Scores tab is initially blank. You must import the LOD scores for the linkage export. See <u>Importing LOD Scores</u>.
- d. Optionally, to rerun the linkage export, right-click on the selected export, and on the context menu that opens, click Open Export.
  - i. The Specify Linkage Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See <u>Creating and Running a Linkage Export</u>.

# **Importing LOD Scores**

After you run a linkage export, the LOD Scores tab is initially blank for a selected export. You must import the LOD scores for the linkage export to populate the tab.

### To import LOD scores

1. On the Analysis window, right-click on the linkage export for which you are importing the LOD scores, and on the context menu that opens, click Import LOD scores. The LOD Score Import Settings dialog box opens.

Figure 4-13: LOD Score Import Settings dialog box



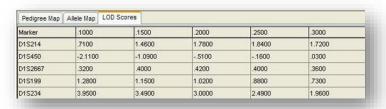
2. Click Browse to open the Open dialog box and browse to and select the LOD Score text files for the linkage export.



Remember, you loaded the files that were created by the linkage export into your analysis software to produce these LOD files.

3. Click Import. The LOD scores for each marker are displayed on the LOD Scores tab on the Analysis window for the selected linkage export.

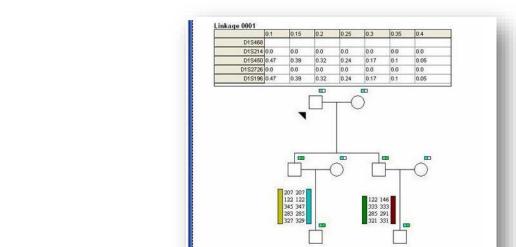
Figure 4-14: LOD Scores tab



# Displaying LOD Scores on a Pedigree

In addition to displaying LOD scores on the LOD Scores tab on the Analysis window, you can display LOD scores on a pedigree. To display the LOD scores on a pedigree, note the following:

- 1. You must load the files that were created by the linkage export into your analysis software to produce the needed LOD files, and then you must import these LOD files into Progeny. See Importing LOD Scores.
- 2. When you are drawing the pedigree, you must select the markers that you are to be displayed on the pedigree. See To configure icon markers for a pedigree.
- 3. Show LOD Scores must be selected on the Pedigree tab of the Properties dialog box. See <u>Pedigree</u> Properties.



201 207

Figure 4-15: Displaying LOD scores on a pedigree

# Deleting a Linkage Export

- 1. You can delete a linkage export by doing one of the following:
  - a. Right-clicking on the linkage export in the Analysis window, and on the context menu that opens, clicking Delete Export.
  - b. Selecting the linkage export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 5 Family Based Associations

Progeny Lab provides all the functionality needed for managing genetic linkage studies, including managing family-based associations. Managing family-based associations consists of creating and running family-based association exports, saving family-based association export formats for running on an as-needed basis, and deleting family-based association exports.

This chapter covers the following topics:

- Creating and Running a Family-Based Association Export
- Saving and Loading a Family Based Association Export Format
- Viewing and Rerunning a Family-Based Association Export
- Deleting a Family-Based Association Export

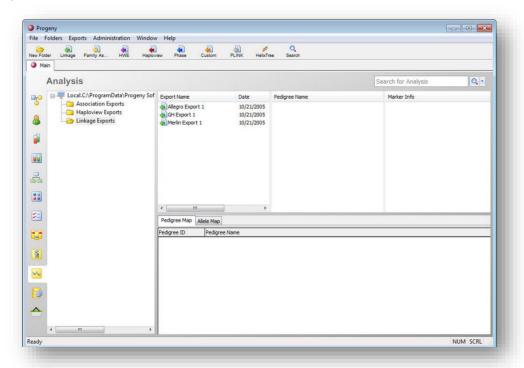
# Creating and Running a Family-Based Association Export

You can export FBAT and QTDT association formats in Progeny Lab. You must first create the association export, and then you can run the association export.

## To create a family-based association export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 5-1: Analysis window



2. On the Analysis window, select the folder in which to the association export is to be saved.

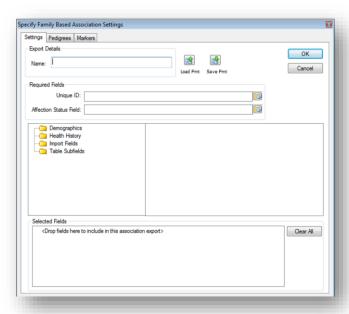


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the Family Association Family Association. The Specify Family Based Association Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary association settings on all three tabs of the dialog box. See:
  - a. Specify Family Based Association Settings dialog box, Settings tab
  - b. Specify Family Based Association Settings dialog box, Pedigrees tab
  - c. Specify Family Based Association Settings dialog box, Markers tab
- 5. After you specify the necessary association settings on all three tabs, return to the Settings tab, and then click OK on the tab.
- 6. Continue to... To run a family-based association export

## Specify Family Based Association Settings dialog box, Settings tab

Figure 5-2: Specify Family Based Association Settings dialog box, Settings tab



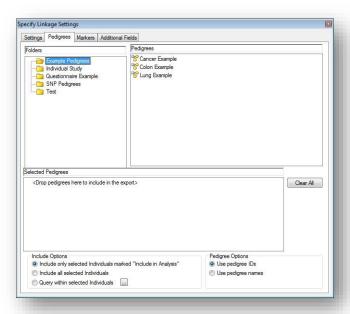
1. Specify the name for the association export and the required fields for the export.

Option	Description	
Export Details		
Name	Name or description of the association export.	
Required Fields		
Unique ID	Click the Fields button to the right of the Unique ID field to open the Select Field dialog box and select the field that represents the unique ID for everyone that is to be included in the association export.	
Affection Status Field	Click the Fields button to the right of the Affection Status Field to open the Select Field dialog box and select the field that represents the affected status for everyone that is to be included in the association export.	

- 2. Select the fields that are to be included in the association export by doing the following:
  - a. In the Folders (middle left) pane of the dialog box, select the folder that contains the individual data fields that are to be included in the association export.
  - b. In the Fields pane (middle right) pane of the dialog, select the fields (CTRL-click to select multiple fields) that are to be included in the association export.
  - c. Drag the selected fields to the Selected fields (bottom) pane of the dialog box.
- 3. Continue specifying any other family-based export settings as needed; otherwise, return to the Settings tab, click OK, and then continue to... To run a family-based association export.

#### Specify Family Based Association Settings dialog box, Pedigrees tab

Figure 5-3: Specify Family Based Association Settings dialog box, Pedigrees tab



- 1. In the Folders (top left) pane of the dialog box, select the folder that contains the pedigrees for which the linkage export is being created.
- 2. In the Pedigrees (top right) pane of the dialog, select the pedigree (CTRL-click to select multiple pedigrees) for which the linkage export is being created.
- 3. Drag the selected pedigrees to the Selected Pedigrees (bottom) pane of the dialog box.
- 4. Select one of the following:
  - To include only those individuals in a pedigree who have been specifically marked for inclusion in an analysis, select Include only selected individuals marked "Include in Analysis." See <u>To select individuals in a pedigree for inclusion in an analysis</u>
  - To include all individuals in all the selected pedigrees are included in the linkage export, select include all selected individuals.
  - c. To include only those individuals in the export linkage who meet specific criteria, select Query within selected individuals, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.



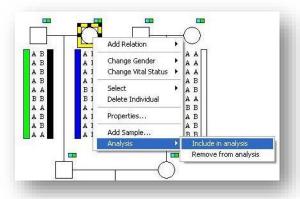
#### For detailed information about defining a query, see Database Queries and **Searches**

- 5. Select one of the following:
  - a. Use Pedigree IDs—Converts a text-based pedigree name into a numerical ID.
  - b. Use pedigree names—Use pedigree names as-is.
- 6. Continue specifying any other family-based export settings as needed; otherwise, return to the Settings tab, click OK, and then continue to... To run a family-based association export

## To select individuals in a pedigree for inclusion in an analysis

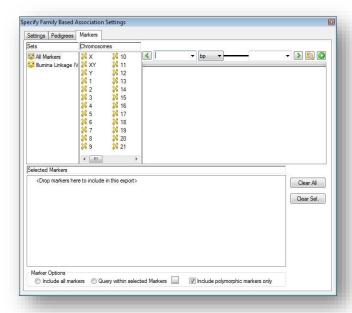
If you select the option Include only selected individuals marked "Include in Analysis," then you must open each affected pedigree, and manually select the appropriate individuals. You can select a single individual, or CTRL-click to select multiple individuals, and then right click on any selected individual, and on the context menu that opens, click Include in analysis.

Figure 5-4: Selecting individuals for analysis



#### Specify Family Based Association Settings dialog box, Markers tab

Figure 5-5: Specify Family Based Association Settings dialog box, Markers tab



#### 1. Do one of the following:

- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See Searching for a Marker.

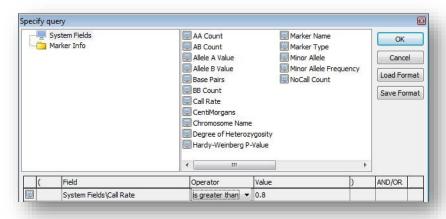
- 2. Select one of the following:
  - a. To include all the markers in the Selected Markers pane in the linkage export, select include all markers.
  - b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

For detailed information about defining a query, see <u>Database Queries and Searches</u>



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

Figure 5-6: Additional marker filtering

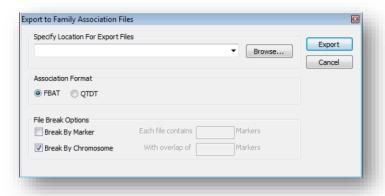


- 3. Optionally, select Include polymorphic markers only to further filter the selected markers and export only those markers that have at least one heterozygous genotype.
- 4. Continue specifying any other family-based export settings as needed; otherwise, return to the Settings tab, click OK, and then continue below.

## To run a family-based association export

After you specify the necessary export settings on all three tabs, you must return to the Settings tab, and then click OK on the tab. The Export to Family Association Files dialog box opens.

Figure 5-7: Export to Family Association Files dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the association export files are to be stored.



If you have previously run association exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

- 2. Select the appropriate association format.
- 3. Select one or both file break options.
  - a. Break by Marker Indicate how many markers are to be contained in each linkage file. If you select this option, you must also indicate the number of overlapping markers in each file.
  - b. Break by Chromosome Creates a new file for each chromosome that is exported.
- 4. Click Export. A dialog box opens, indicating the status of the association export.
- 5. When the export is complete, click Close to close the dialog box and return to the Analysis window.

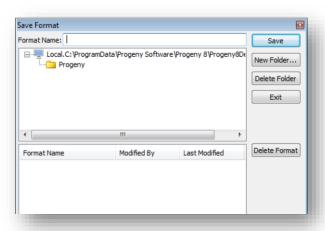
# Saving and Loading a Family Based Association Export Format

After you have set up a family-based association export format, you can save the format. You can then load this saved format and run it on as-needed basis.

## To save a family-based association export format

- 1. Set up the family-based association export format but do not run the format See <u>Creating and Running a Family-Based Association Export.</u>
- 2. Click the Save Format Save Fmt button on the Specify Family Based Association Settings dialog box.
  - a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 5-8: Save Format dialog box



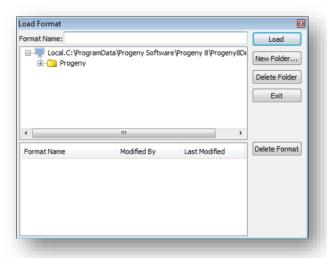
- 3. Enter a name for the association export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder. Click Save.
- 4. Click Save on the Save Format dialog box. You can then load this saved format and run it on as-needed basis. See To load a family-based association export format

## To load a family-based association export format

You can a retrieve a saved family-based association export format and run it on as-is needed based.

- 1. Open the Specify Family Based Association Settings dialog box. (See <u>To create a family-based association export</u> steps 1-3)
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 5-9: Load Format dialog box



- 3. Open the folder that contains the association export format that you are loading, select the format, and then click Load.
  - a. The Specify Family Based Association Export Settings dialog box is refreshed with the loaded format. You can modify the format as needed (for example, modify the information on the Pedigrees tab), and export the association format.

# Viewing and Rerunning a Family-Based Association Export

You can view information about any family-based association exports that were previously run on the Analysis window, and you can rerun any association export as necessary.

## To view and rerun a family-based association export

- 1. On the Analysis window, select the association export that you want to view.
  - a. Two tabs open in the bottom pane of the window—Pedigree Map and Allele Map. By default, the Pedigree Map tab is the active tab.
- 2. Open and view the information on each tab as necessary.
  - a. The Pedigree Map tab displays the mapping of the original pedigree name to the pedigree ID generated for the export file. These values are not editable; however, you can sort this information by clicking the heading for either the Pedigree ID or the Pedigree Name.

Figure 5-10: Pedigree Map tab



b. The Allele Map tab displays the mapping of the original calls for each marker to the allele number generated for the export file. These values are not editable; however, you can sort the data by clicking the heading for either the Maker, Allele, or Call.

Figure 5-11: Allele Map tab





You can also right-click on either of these tabs and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 3. Optionally, to rerun the association export, right-click on the selected export, and on the context menu that opens, click Open Export.
  - a. The Specify Family Based Association Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See Creating and Running a Family-Based Association Export.

# **Deleting a Family-Based Association Export**

You can delete a family-based association export by doing one of the following:

- 1. Right-clicking on the association export in the Analysis window, and on the context menu that opens, clicking Delete Export.
- 2. Selecting the association export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 6 Hardy Weinberg Test

Progeny Lab provides all the functionality needed for creating Hardy Weinberg tests and working with the results.

This chapter covers the following topics:

- Creating and Running a Hardy Weinberg Export
- Saving and Loading a Hardy Weinberg Export Format
- Viewing and Rerunning a Hardy Weinberg Export
- <u>Deleting a Hardy Weinberg Export</u>

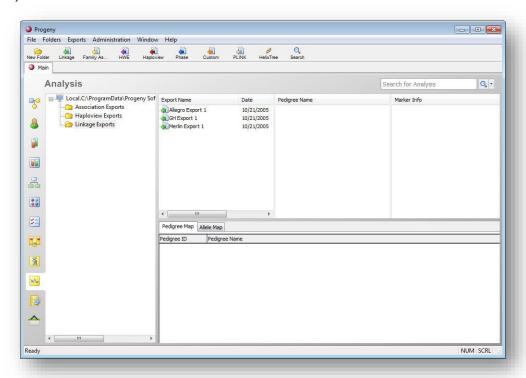
# Creating and Running a Hardy Weinberg Export

Progeny Lab provides all the functionality needed for creating a Hardy Weinberg export and working with the results. You must first create the export, and then you can run the export.

## To create a Hardy Weinberg export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 6-1: Analysis window



2. On the Analysis window, select the folder in which to the association export is to be saved.



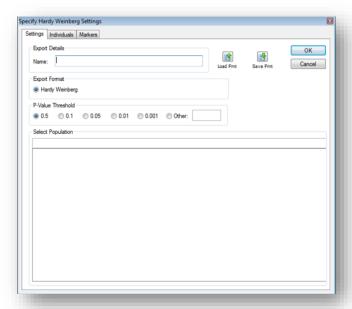
You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)



- 3. On the window toolbar, click the Hardy Weinberg button
  - a. The Specify Hardy Weinberg Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary association settings on all three tabs of the dialog box. See:
  - a. Specify Hardy Weinberg Settings dialog box, Settings tab
  - b. Specify Hardy Weinberg Settings dialog box, Individuals tab
  - c. Specify Hardy Weinberg Settings dialog box, Markers tab
- 5. After you specify the necessary settings on all three tabs, return to the Settings tab then click OK on the tab.
- 6. Continue To run a Hardy Weinberg export

Specify Hardy Weinberg Settings dialog box, Settings tab

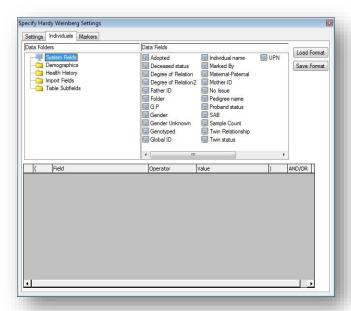
Figure 6-2: Specify Hardy Weinberg Settings dialog box, Settings tab



- 1. Specify the name for the Hardy Weinberg export.
- 2. Select from one of the preset values for the p-value threshold, or click Other, and then enter the p-value threshold.
- 3. Continue specifying any other Hardy Weinberg export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a Hardy Weinberg export</u>

### Specify Hardy Weinberg Settings dialog box, Individuals tab

Figure 6-3: Specify Hardy Weinberg Settings dialog box, Individuals tab



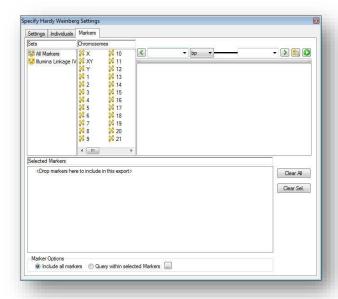
- 1. Define a query to select those individuals that are to be included in your Hardy Weinberg test.
- 2. Continue specifying any other Hardy Weinberg export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a Hardy Weinberg export



For detailed information about defining a query, see <u>Database Queries and</u> <u>Searches</u>

#### Specify Hardy Weinberg Settings dialog box, Markers tab

Figure 6-4: Specify Hardy Weinberg Settings dialog box, Markers tab



#### 1. Do one of the following:

- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See <u>Searching for a Marker</u>

#### 2. Select one of the following:

- a. To include all the markers in the Selected Markers pane in the linkage export, select Include all markers.
- b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

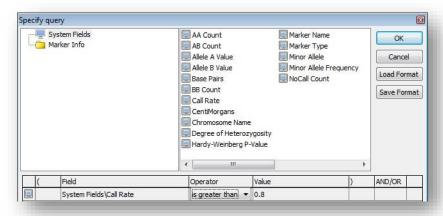


For detailed information about defining a query, see <u>Database Queries and</u> Searches



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

Figure 6-5: Additional marker filtering



3. Continue specifying any other Hardy Weinberg export settings as needed; otherwise, return to the Settings tab, click OK, and then continue.

## To run a Hardy Weinberg export

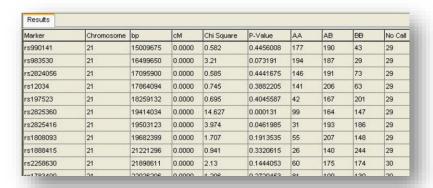
- 1. After you specify the necessary export settings on all three tabs, you must return to the Settings tab, and then click OK on the tab.
  - a. An Export Status dialog box opens, indicating the progress of the export.
- 2. After the export is completed, click Close to close the Export Status dialog box and return to the Analysis window.
  - a. The resulting is displayed on the Results tab in the bottom pane of the Analysis window.



Remember, the results might not contain all the markers that you selected. If you specified a lower p-value threshold, then the higher p-value results are not included.

Column	Description
Marker	The name of the marker.
Chromosome	The name of the chromosome.
bp	The base pair information for the chromosome.
сМ	The cM information for the marker.
Chi Square	The calculated Chi Square value for the marker.
P-Value	The corresponding p-value for the marker.
AA	The number of AA calls in the Hardy Weinberg test.
AB	The number of AB calls in the Hardy Weinberg test.
ВВ	The number of BB calls in the Hardy Weinberg test.
No Calls	The number of No Calls in the Hardy Weinberg test.

Figure 6-6: Results tab



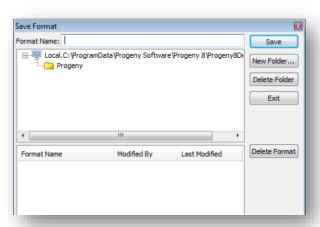
## Saving and Loading a Hardy Weinberg Export Format

After you have set up a Hardy Weinberg export format, you can save the format. You can then load this saved format and run it on as-needed basis.

### To save a Hardy Weinberg export format

- 1. Set up the Hardy Weinberg export format but do not run the format. See <u>Creating and Running a Hardy Weinberg Export</u>
- 2. Click the Save Format Save Fmt button on the Specify Hardy Weinberg Settings dialog box.
  - a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 6-7: Save Format dialog box



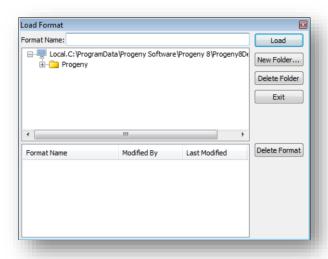
- 3. Enter a name for the Hardy Weinberg export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
  - c. Enter the name of the new folder in which to save to format.
  - d. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
  - e. Click Save.
- 4. Click Save on the Save Format dialog box. You can then load this saved format and run it on as-needed basis.

### To load a Hardy Weinberg export format

You can a retrieve a saved Hardy Weinberg export format and run it on as-is needed based.

- 1. Open the Specify Hardy Weinberg Settings dialog box. See Creating and Running a Hardy Weinberg Export
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 6-8: Load Format dialog box



- 3. Open the folder that contains the Hardy Weinberg export format that you are loading, select the format, and then click Load.
- 4. The Specify Hardy Weinberg Settings dialog box is refreshed with the loaded format.
  - a. You can modify the format as needed (for example, modify the information on the Markers tab), and export the format. See <u>Creating and Running a Hardy Weinberg Export</u>

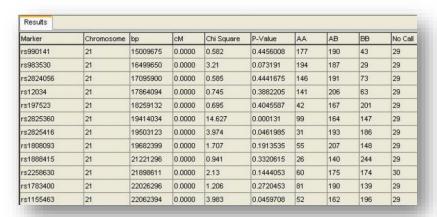
## Viewing and Rerunning a Hardy Weinberg Export

You can view information about any Hardy Weinberg exports that were previously run on the Analysis window, and you can rerun any Hardy Weinberg export as necessary.

### To view and rerun a Hardy Weinberg export

- 1. On the Analysis window, select the Hardy Weinberg export that you want to view.
  - a. A single tab the Results tab opens in the bottom pane of the window.

Figure 6-9: Results tab





You can also right-click on the tab and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 2. Optionally, to rerun the Hardy Weinberg export, right-click on the selected export, and on the context menu that opens, click Open Export.
  - a. The Specify Hardy Weinberg Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See <u>Creating and Running a Hardy</u> <u>Weinberg Export</u>

# Deleting a Hardy Weinberg Export

You can delete a Hardy Weinberg export by doing one of the following:

- 1. Right-clicking on the export in the Analysis window, and on the context menu that opens, clicking Delete Export.
- 2. Selecting the export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 7 Haploview Exports

Progeny Lab provides all the functionality needed for creating Haploview exports and working with the results.

This chapter covers the following topics:

- **Creating and Running a Haploview Export**
- Saving and Loading a Haploview Export Format
- Viewing and Rerunning a Haploview Export
- **Deleting a Haploview Export**



To create and run Haploview exports, Haploview must be installed locally.

# Creating and Running a Haploview Export

Progeny Lab provides all the functionality needed for creating Haploview exports and working with the results. You must first create the export, and then you can run the export.

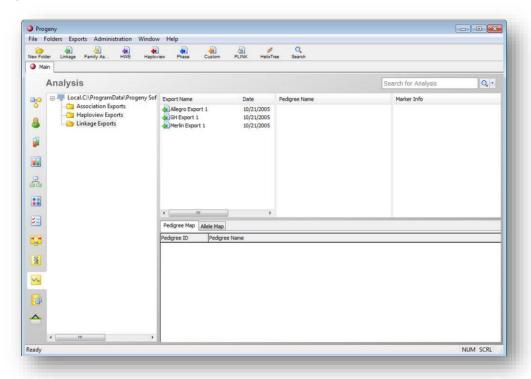


To create and run Haploview exports, Haploview must be installed locally.

### To create a Haploview export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 7-1: Analysis window



2. On the Analysis window, select the folder in which to the Haploview export is to be saved.

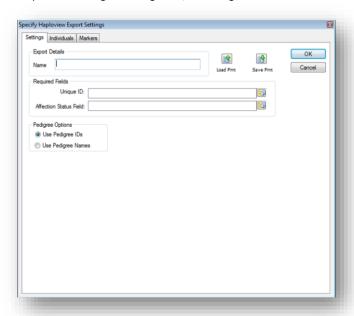


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the Haploview Haploview button. The Specify Haploview Export Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary association settings on all three tabs of the dialog box. See:
  - a. Specify Haploview Export Settings dialog box, Settings tab
  - b. Specify Haploview Export Settings dialog box, Individuals tab
  - c. Specify Haploview Export Settings dialog box, Markers tab
- 5. After you specify the necessary export settings on all three tabs, return to the Settings tab and click OK on the tab. Continue To run a Haploview export

### Specify Haploview Export Settings dialog box, Settings tab

Figure 7-2: Specify Haploview Export Settings dialog box, Settings tab



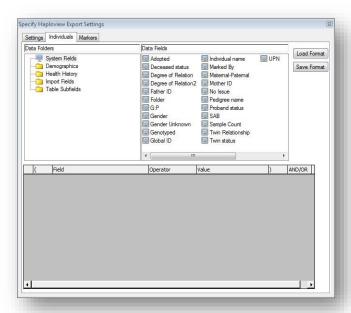
1. Specify the settings for the Haploview export.

Option	Description
Name	Name or description of the Haploview export.
Unique ID	Click the Fields button to the right of the Unique ID field to open the Select Field dialog box and select the field that represents the unique ID for everyone that is to be included in the Haploview export.
Affection Status Field	Click the Fields button to the right of the Affection Status Field to open the Select Field dialog box and select the field that represents the affected status for everyone that is to be included in the Haploview export.
Use Pedigree IDs	Converts a text-based pedigree name into a numerical ID.
Use pedigree names	Use pedigree names as-is.

2. Continue specifying any other Haploview export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a Haploview export</u>

#### Specify Haploview Export Settings dialog box, Individuals tab

Figure 7-3: Specify Haploview Export Settings dialog box, Individuals tab



1. Define a query to select those individuals that are to be included in your Haploview export.

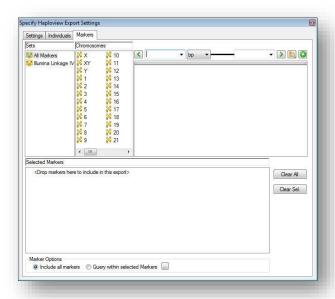


For detailed information about defining a query, see <u>Database Queries and</u> Searches

2. Continue specifying any other Haploview export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a Haploview export</u>

#### Specify Haploview Export Settings dialog box, Markers tab

Figure 7-4: Specify Haploview Export Settings dialog box, Markers tab



#### 1. Do one of the following:

- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See Searching for a Marker

#### 2. Select one of the following:

- a. To include all the markers in the Selected Markers pane in the linkage export, select Include all markers.
- b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

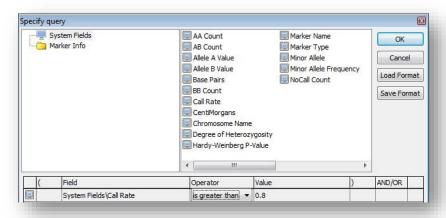


For detailed information about defining a query, see <u>Database Queries and</u> Searches



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

Figure 7-5: Additional marker filtering

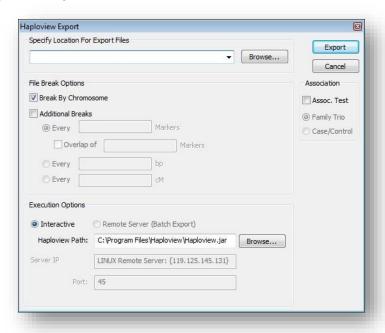


3. Continue specifying any other Haploview export settings as needed; otherwise, return to the Settings tab, click OK, and then continue.

### To run a Haploview export

After you specify the necessary export settings on all three tabs, you must return to the Settings tab, and then click OK on the tab. The Haploview Export dialog box opens.

Figure 7-6: Haploview Export dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the Haploview export files are to be stored.



If you have previously run Haploview exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

2. Optionally to select an association format, select Association Test, and then select Family Trios or Case/Controls.

- 3. Select one or both file break options.
  - a. Break by Chromosome—Creates a new file for each chromosome that is exported.
  - b. Additional Breaks
    - i. Every [] Markers—Indicates how many markers are to be contained in each export file. If you select this option, you must also indicate the number of overlapping markers in each
    - ii. Every [] bp—Indicates how many base pairs are to be included in each file.
    - iii. Every [] cM—Indicates how many centimorgans are to be included in each file.



The break options can overlap. For example, select to Break by Chromosome and then by every 1000000 bp within the chromosome.

- 4. Only the Interactive execution is currently available. The default path to the Haploview executable is displayed in the Haploview Path field. You can leave this location as-is, or you can click Browse to open the Open the dialog box and browse to and select a different location for the executable.
- 5. Click Export. An Export to Haploview dialog box opens, indicating the status of the Haploview export.
- 6. When the export is complete, click Close to close the dialog box and return to the Analysis window.
  - a. After Haploview has completed the necessary calculations, a Haploview window opens showing the result of the calculations.

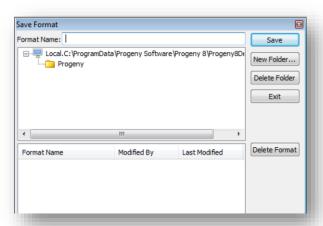
## Saving and Loading a Haploview Export Format

After you have set up a Haploview export format, you can save the format. You can then load this saved format and run it on as-needed basis.

### To save a Haploview export format

- 1. Set up the Haploview export format but do not run the format See Creating and Running a Haploview **Export**
- 2. Click the Save Format Save Fmt button on the Specify Haploview Export Settings dialog box.
  - a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format.

Figure 7-7: Save Format dialog box



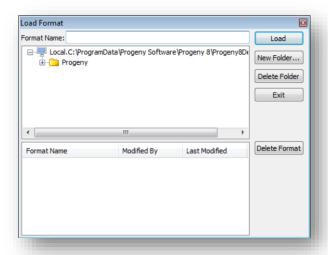
- 3. Enter a name for the Haploview export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
    - iii. Click Save.
- 4. Click Save on the Save Format dialog box. You can then load this saved format and run it on as-needed basis.

### To load a Haploview export format

You can a retrieve a saved Haploview export format and run it on as-is needed based.

- 1. Open the Specify Haploview Export Settings dialog box. See To create a Haploview export
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 7-8: Load Format dialog box



- 3. Open the folder that contains the Haploview export format that you are loading, select the format, and then click Load.
- 4. The Specify Haploview Export Settings dialog box is refreshed with the loaded format.
  - a. You can modify the format as needed (for example, modify the information on the Markers tab), and export the format. See <u>Creating and Running a Haploview Export</u>

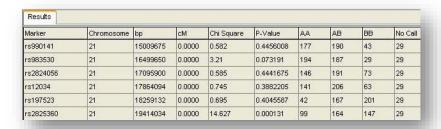
## Viewing and Rerunning a Haploview Export

You can view information about any Haploview exports that were previously run on the Analysis window, and you can rerun any Haploview export as necessary.

### To view and rerun a Haploview export

- 1. On the Analysis window, select the Haploview export that you want to view.
  - a. A single tab the Results tab opens in the bottom pane of the window.

Figure 7-9: Results tab





You can also right-click on the tab and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 2. Optionally, to rerun the Haploview export, right-click on the selected export, and on the context menu that opens, click Open Export.
  - a. The Specify Haploview Export Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See <a href="Creating and Running a Haploview Export">Creating and Running a Haploview Export</a>

# Deleting a Haploview Export

You can delete a Haploview export by doing one of the following:

- 1. Right-clicking on the export in the Analysis window, and on the context menu that opens, clicking Delete Export.
- 2. Selecting the export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 8 Phase Exports

Progeny Lab provides all the functionality needed for creating phase exports and working with the results.

This chapter covers the following topics:

- Creating and Running a Phase Export
- Saving and Loading a Phase Export Format
- Viewing and Rerunning a Phase Export
- Deleting a Phase Export

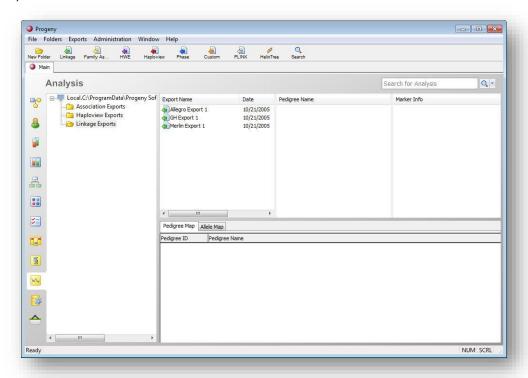
### Creating and Running a Phase Export

Progeny Lab provides all the functionality needed for creating phase exports and working with the results. You must first create the phase export, and then you can run the export.

### To create a phase export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 8-1: Analysis window



2. On the Analysis window, select the folder in which to the phase export is to be saved.

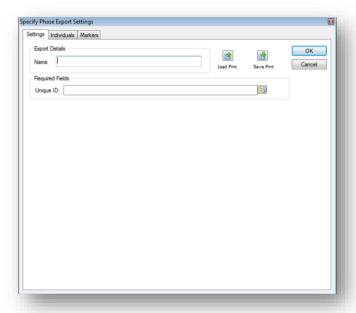


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the Phase button. The Specify Phase Export Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary export settings on all three tabs of the dialog box. See:
- a. Specify Phase Export Settings dialog box, Settings tab
- b. Specify Phase Export Settings dialog box, Individuals tab
- c. Specify Phase Export Settings dialog box, Markers tab
- 5. After you specify the necessary export settings on all three tabs, return to the Settings tab, and then click OK on the tab.
- 6. Continue To run a phase export

Specify Phase Export Settings dialog box, Settings tab

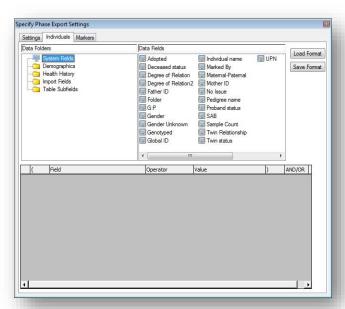
Figure 8-2: Specify Phase Export Settings dialog box, Settings tab



- 1. Specify the name for the phase export
- 2. Click the Fields button to the right of the Unique ID field to open the Select Field dialog box and select the field that represents the unique ID for everyone that is to be included in the phase export.
- 3. Continue specifying any other phase export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a phase export

#### Specify Phase Export Settings dialog box, Individuals tab

Figure 8-3: Specify Phase Export Settings dialog box, Individuals tab



1. Define a query to select those individuals that are to be included in your phase export.

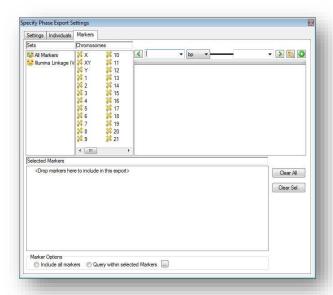


For detailed information about defining a query, see <u>Database Queries and</u> Searches

2. Continue specifying any other phase export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a phase export</u>

#### Specify Phase Export Settings dialog box, Markers tab

Figure 8-4: Specify Phase Export Settings dialog box, Markers tab



- 1. Do one of the following:
- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See <u>Searching for a Marker</u>

- 2. Select one of the following:
- a. To include all the markers in the Selected Markers pane in the linkage export, select Include all markers.
- b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

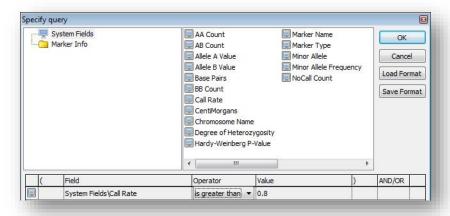


For detailed information about defining a query, see <u>Database Queries and</u> Searches



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

Figure 8-5: Additional marker filtering

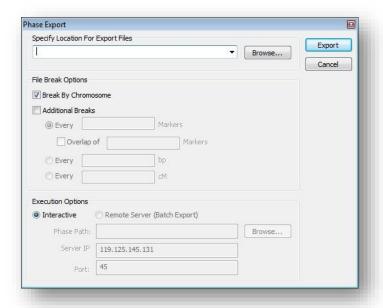


3. Continue specifying any other phase export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a phase export below.

### To run a phase export

After you specify the necessary export settings on all three tabs, you must return to the Settings tab, and then click OK on the tab. The Phase Export dialog box opens.

Figure 8-6: Phase Export dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the phase export is to be stored.



If you have previously run phase exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

- 2. Select one or both file break options.
  - a. Break by Chromosome Creates a new file for each chromosome that is exported.
  - b. Additional Breaks
    - i. Every [] Markers—Indicates how many markers are to be contained in each export file. If you select this option, you must also indicate the number of overlapping markers in each file.
    - ii. Every [] bp—Indicates how many base pairs are to be included in each file.
    - iii. Every [] cM—Indicates how many centimorgans are to be included in each file.



The break options can overlap. For example, select to Break by Chromosome and then by every 1000000 bp within the chromosome.



Execution options are currently not applicable.

3. Click Export. An Export to Phase dialog box opens, indicating the status of the phase export. When the export is complete, click Close to close the dialog box and return to the Analysis window.

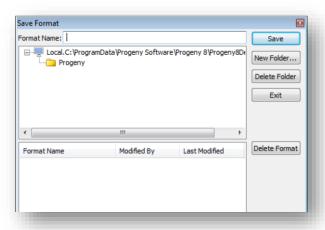
## Saving and Loading a Phase Export Format

After you have set up a phase export format, you can save the format. You can then load this saved format and run it on as-needed basis.

### To save a phase export format

- 1. Set up the phase export format but do not run the format See Creating and Running a Phase Export
- 2. Click the Save Format Save Fmt button on the Specify Phase Export Settings dialog box.
  - a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 8-7: Save Format dialog box



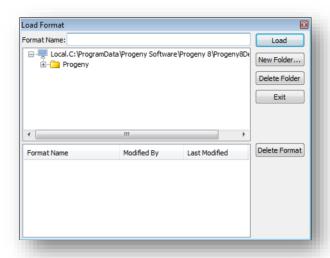
- 3. Enter a name for the phase export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
    - iii. Click Save.
- 4. Click Save on the Save Format dialog box. You can then load this saved format and run it on as-needed basis.

### To load a phase export format

You can a retrieve a saved phase export format and run it on as-is needed based.

- 1. Open the Specify Phase Export Settings dialog box. See To create a phase export
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 8-8: Load Format dialog box



- 3. Open the folder that contains the phase export format that you are loading, select the format, and then click Load.
  - a. The Specify Phase Export Settings dialog box is refreshed with the loaded format. You can modify the format as needed (for example, modify the information on the Markers tab), and export the format. See Creating and Running a Phase Export

## Viewing and Rerunning a Phase Export

You can view information about any phase exports that were previously run on the Analysis window, and you can rerun any phase export as necessary.

### To view and rerun a phase export

- 1. On the Analysis window, select the phase export that you want to view.
  - a. A single tab the Results tab opens in the bottom pane of the window.



You can also right-click on the tab and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 2. Optionally, to rerun the phase export, right-click on the selected export, and on the context menu that opens, click Open Export.
  - a. The Specify Phase Export Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See <u>Creating and Running a Phase Export</u>

## **Deleting a Phase Export**

You can delete a phase export by doing one of the following:

- 1. Right-clicking on the export in the Analysis window, and on the context menu that opens, clicking Delete Export.
- 2. Selecting the export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 9 Custom Genotype Exports

Progeny Lab provides all the functionality needed for creating custom genotype exports and working with the results.

This chapter covers the following topics:

- Creating and Running a Custom Genotype Export
- Saving and Loading a Custom Genotype Export Format
- Viewing and Rerunning a Custom Genotype Export
- Deleting a Custom Genotype Export

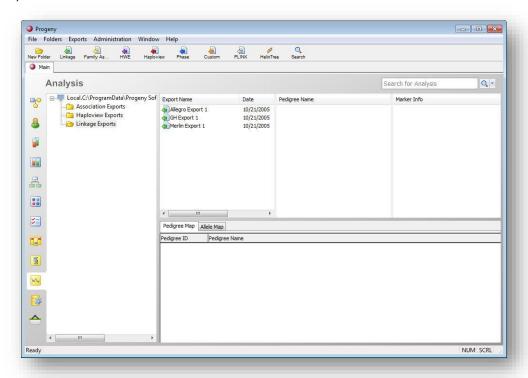
### Creating and Running a Custom Genotype Export

Progeny Lab provides all the functionality needed for creating custom genotype exports and working with the results. You must first create the custom export, and then you can run the export.

### To create a custom genotype export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 9-1: Analysis window



2. On the Analysis window, select the folder in which to the custom export is to be saved.

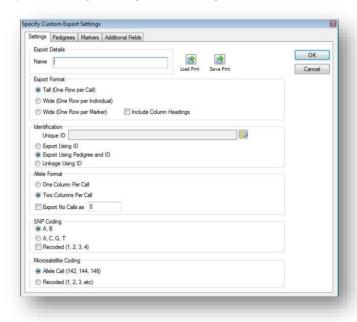


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the Custom button. The Specify Custom Export Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary export settings on all four tabs of the dialog box. See:
- a. Specify Custom Export Settings dialog box, Settings tab
- b. Specify Custom Export Settings dialog box, Pedigrees tab
- c. Specify Custom Export Settings dialog box, Markers tab
- d. Specify Custom Export Settings dialog box, Additional Fields tab
- 5. After you specify the necessary export settings on all four tabs, return to the Settings tab, and then click OK on the tab.
- 6. Continue To run a custom genotype export

### Specify Custom Export Settings dialog box, Settings tab

Figure 9-2: Specify Custom Export Settings dialog box, Settings tab



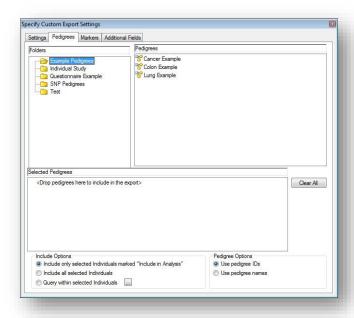
1. Specify the settings for the custom export.

Option	Description	
Export Details		
Name	Name or description of the custom export.	
Export Format		
Tall (One Row per Call)	Unique ID, Marker Name, Allele 1, and Allele 2	
Wide (One Row per Individual)	Individuals in rows and markers in columns (Two columns per call)	
Wide (One Row per Marker)	Markers in rows and individuals in columns (Two columns per call)	
Affection Status Field	Click the Fields button to the right of the Affection Status Field to open the Select Field dialog box and select the field that represents the affected status for everyone that is to be included in the PLINK export.	
	Pedigree Options	
Use Pedigree IDs	Converts a text-based pedigree name into a numerical ID.	
Use pedigree names	Use pedigree names as-is.	
	Identification	
	Inique ID field to open the Select Field dialog box and select the identifier, such as the exported, then select one of the identification options.	
Export Using ID	Use the selected ID to identify the individuals being exported.	
Export Using Pedigree and UPN	Use the pedigree and the selected ID to identify the individuals being exported.	
Linkage Using ID	When you select an ID, the same ID field is exported to specify the Mother and Father.	
Allele Format		
One Column Per Call	For example, AB is shown in a single column.	
Two Columns Per Call	For example, A is shown in one column and B is shown in the second column.	
Export No Calls as	Optional. The default value is 0, but you can enter another value.	
	SNP Coding	
A, B	Show alleles as A or B.	
A, C, G, T	Show alleles as A, C, G, or T.  Note: If you select A, C, G, or T, you can select Recoded (1, 2, 3, 4) and show the alleles as 1, 2, 3, or 4 instead of A, C, G, or T.	
Microsatellite Coding		
Allele Call	Use the actual allele call in the export.	
Recoded (1, 2, 3, etc.)	Recode the allele call to 1, 2, 3, and so on in the export.	

2. Continue specifying any other custom export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a custom genotype export

### Specify Custom Export Settings dialog box, Pedigrees tab

Figure 9-3: Specify Custom Export Settings dialog box, Pedigrees tab



- 1. In the Folders (top left) pane of the dialog box, select the folder that contains the pedigrees for which the linkage export is being created.
- 2. In the Pedigrees (top right) pane of the dialog, select the pedigree (CTRL-click to select multiple pedigrees) for which the linkage export is being created.
- 3. Drag the selected pedigrees to the Selected Pedigrees (bottom) pane of the dialog box.
- 4. Select one of the following:
- 5. To include only those individuals in a pedigree who have been specifically marked for inclusion in an analysis, select Include only selected individuals marked "Include in Analysis." See <u>To select individuals in a pedigree for inclusion in an analysis</u>
- 6. To include all individuals in all the selected pedigrees are included in the linkage export, select Include all selected individuals.
- 7. To include only those individuals in the export linkage who meet specific criteria, select Query within selected individuals, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.



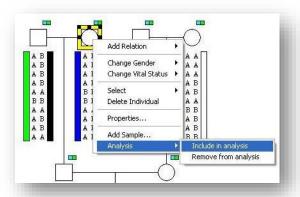
#### For detailed information about defining a query, see <u>Database Queries and</u> Searches

- 8. Select one of the following:
  - a. Use Pedigree IDs—Converts a text-based pedigree name into a numerical ID.
  - b. Use pedigree names—Use pedigree names as-is.
- 9. Continue specifying any other custom export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a custom genotype export

#### To select individuals in a pedigree for inclusion in an analysis

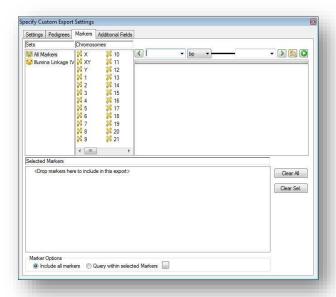
If you select the option Include only selected individuals marked "Include in Analysis," then you must open each affected pedigree, and manually select the appropriate individuals. You can select a single individual, or CTRL-click to select multiple individuals, and then right click on any selected individual, and on the context menu that opens, click Include in analysis.

Figure 9-4: Selecting individuals for analysis



#### Specify Custom Export Settings dialog box, Markers tab

Figure 9-5: Specify Custom Export Settings dialog box, Markers tab



- 1. Do one of the following:
- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See <u>Searching for a Marker</u>

- 2. Select one of the following:
- a. To include all the markers in the Selected Markers pane in the linkage export, select Include all markers.
- b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

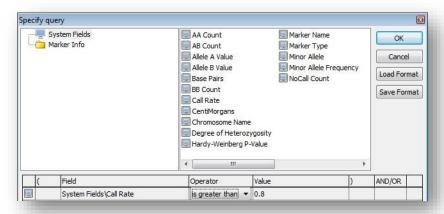


For detailed information about defining a query, see <u>Database Queries and</u> Searches



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

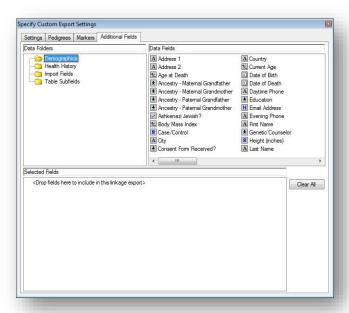
Figure 9-6: Additional marker filtering



3. Continue specifying any other custom export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a custom genotype export

#### Specify Custom Export Settings dialog box, Additional Fields tab

Figure 9-7: Specify Custom Export Settings dialog box, Additional Fields tab

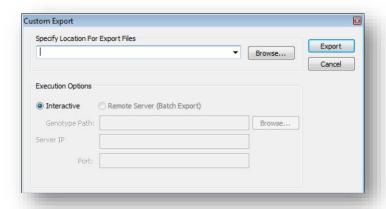


- 1. Specify additional individual level database fields that are to be included in the custom genotype export.
- 2. Continue specifying any other custom export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a custom genotype export

## To run a custom genotype export

After you specify the necessary export settings on all fours tabs, you must return to the Settings tab, and then click OK on the tab. The Custom Export dialog box opens.

Figure 9-8: Custom Export dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the custom export files are to be stored.



If you have previously run custom exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

Execution options are currently not applicable.

- 2. Click Export. A Custom Export dialog box opens, indicating the status of the custom export.
- 3. When the export is complete, click Close to close the dialog box and return to the Analysis window.

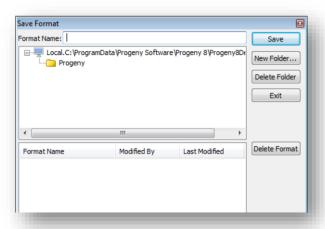
# Saving and Loading a Custom Genotype Export Format

After you have set up a custom genotype export format, you can save the format. You can then load this saved format and run it on as-needed basis.

## To save a custom genotype export format

- 1. Set up the custom genotype export format but do not run the format See <u>Creating and Running a Custom Genotype Export</u>
- 2. Click the Save Format Save Fmt button on the Specify Custom Export Settings dialog box.
- a. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 9-9: Save Format dialog box



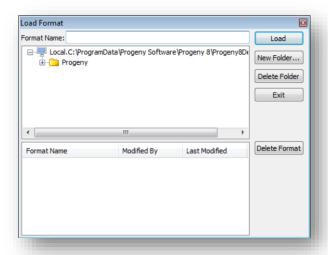
- 3. Enter a name for the custom export format, and then do one of the following:
- a. Select the Progeny folder.
- b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
  - i. Enter the name of the new folder in which to save to format.
  - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
  - iii. Click Save.
- 4. Click Save on the Save Format dialog box.
- 5. You can then load this saved format and run it on as-needed basis.

## To load a custom genotype export format

You can a retrieve a saved custom genotype export format and run it on as-is needed based.

- 1. Open the Specify Custom Export Settings dialog box. See To create a custom genotype export
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 9-10: Load Format dialog box



1. Open the folder that contains the custom export format that you are loading, select the format, and then click Load.

The Specify Custom Export Settings dialog box is refreshed with the loaded format. You can modify the format as needed (for example, modify the information on the Markers tab), and export the format. See <a href="Creating and Running a Custom Genotype Export">Creating and Running a Custom Genotype Export</a>

# Viewing and Rerunning a Custom Genotype Export

You can view information about any custom genotype exports that were previously run on the Analysis window, and you can rerun any custom genotype exports as necessary.

## To view and rerun a custom genotype export

- 1. On the Analysis window, select the phase export that you want to view.
- a. A single tab the Results tab opens in the bottom pane of the window.



You can also right-click on the tab and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 2. Optionally, to rerun the custom export, right-click on the selected export, and on the context menu that opens, click Open Export.
- a. The Specify Custom Export Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See <u>Creating and Running a Custom Genotype</u> <u>Export</u>

## **Deleting a Custom Genotype Export**

You can delete a custom genotype export by doing one of the following:

- Right-clicking on the export in the Analysis window, and on the context menu that opens, clicking Delete Export.
- Selecting the export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 10 PLINK Exports

Progeny Lab provides all the functionality needed for creating PLINK exports and working with the results.

This chapter covers the following topics:

- Creating and Running a PLINK Export
- Saving and Loading a PLINK Export Format
- Viewing and Rerunning a PLINK Export
- Deleting a PLINK Export

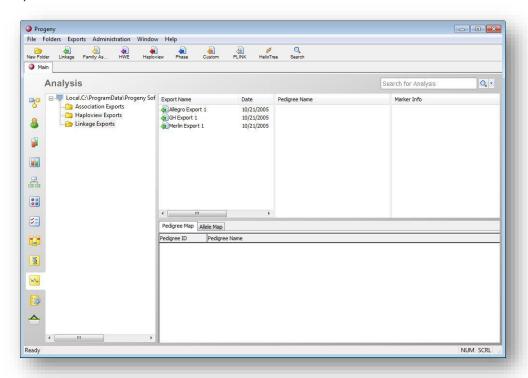
# Creating and Running a PLINK Export

Progeny Lab provides all the functionality needed for creating PLINK exports and working with the results. You must first create the export, and then you can run the export.

## To create a PLINK export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 10-1: Analysis window



2. On the Analysis window, select the folder in which to the phase export is to be saved.

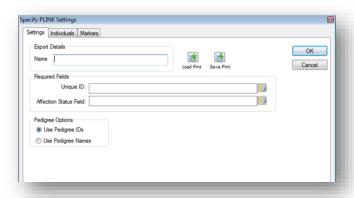


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the PLINK PLINK button. The Specify Phase Export Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary export settings on all three tabs of the dialog box. See:
  - a. Specify PLINK Export Settings dialog box, Settings tab
  - b. Specify PLINK Export Settings dialog box, Individuals tab
  - c. Specify PLINK Export Settings dialog box, Markers tab
- 5. After you specify the necessary export settings on all three tabs, return to the Settings tab, and then click OK on the tab.
- 6. Continue To run a PLINK export

#### Specify PLINK Export Settings dialog box, Settings tab

Figure 10-2: Specify PLINK Export Settings dialog box, Settings tab



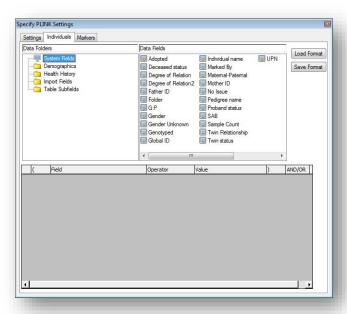
1. Specify the name for the PLINK export, the required fields for the export, and the pedigree options for the export.

Option	Description	
Export Details		
Name	Name or description of the PLINK export.	
Required Fields		
Unique ID	Click the Fields button to the right of the Unique ID field to open the Select Field dialog box and select the field that represents the unique ID for everyone that is to be included in the PLINK export.	
Affection Status Field	Click the Fields button to the right of the Affection Status Field to open the Select Field dialog box and select the field that represents the affected status for everyone that is to be included in the PLINK export.	
Pedigree Options		
Use Pedigree IDs	Converts a text-based pedigree name into a numerical ID.	
Use pedigree names	Use pedigree names as-is.	

2. Continue specifying any other PLINK export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a PLINK export

#### Specify PLINK Export Settings dialog box, Individuals tab

Figure 10-3: Specify Phase Export Settings dialog box, Individuals tab



1. Define a query to select those individuals that are to be included in your Phase export.

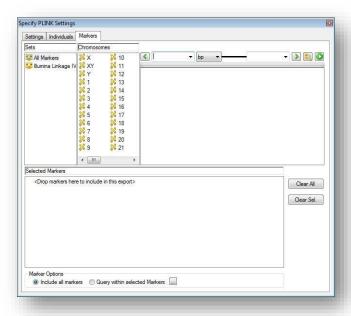


For detailed information about defining a query, see <u>Database Queries and</u> Searches

2. Continue specifying any other PLINK export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a PLINK export</u>

#### Specify PLINK Export Settings dialog box, Markers tab

Figure 10-4: Specify PLINK Export Settings dialog box, Markers tab



#### 1. Do one of the following:

- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See Searching for a Marker

- 2. Select one of the following:
  - a. To include all the markers in the Selected Markers pane in the linkage export, select Include all markers.
  - b. To include only those markers in the Selected Markers pane in the linkage export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

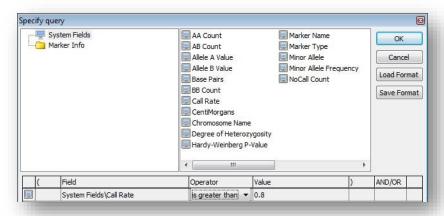


For detailed information about defining a query, see <u>Database Queries and</u> Searches



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

Figure 10-5: Additional marker filtering

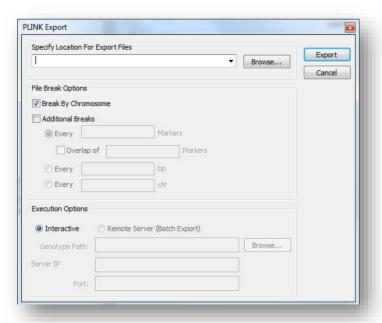


3. Continue specifying any other PLINK export settings as needed; otherwise, return to the Settings tab, click OK, and then continue To run a PLINK export below.

## To run a PLINK export

After you specify the necessary export settings on all three tabs, you must return to the Settings tab, and then click OK on the tab. The PLINK Export dialog box opens.

Figure 10-6: PLINK Export dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the PLINK exports files are to be stored.



If you have previously run PLINK exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

- 2. Select one or both file break options.
  - a. Break by Chromosome Creates a new file for each chromosome that is exported.
  - b. Additional Breaks
    - Every [] Markers Indicates how many markers are to be contained in each export file. If you select this option, you must also indicate the number of overlapping markers in each file.
    - ii. Every [] bp Indicates how many base pairs are to be included in each file.
    - iii. Every [] cM Indicates how many centimorgans are to be included in each file.



The break options can overlap. For example, select to Break by Chromosome and then by every 1000000 bp within the chromosome.

Execution options are currently not applicable.

- 3. Click Export. An Export to PLINK dialog box opens, indicating the status of the phase export.
- 4. When the export is complete, click Close to close the dialog box and return to the Analysis window.

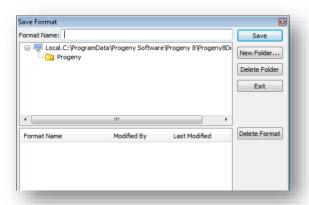
# Saving and Loading a PLINK Export Format

After you have set up a PLINK export format, you can save the format. You can then load this saved format and run it on as-needed basis.

## To save a PLINK export format

- 1. Set up the PLINK export format but do not run the format See Creating and Running a PLINK Export
- 2. Click the Save Format button Save Fmt on the Specify PLINK Export Settings dialog box.
- 3. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 10-7: Save Format dialog box



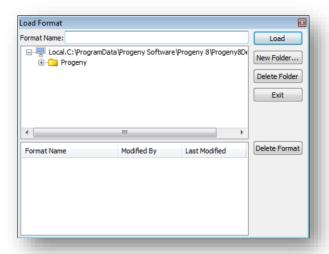
- 4. Enter a name for the PLINK export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder.
    - iii. Click Save.
- 5. Click Save on the Save Format dialog box. You can then load this saved format and run it on as-needed basis. See <u>To load a PLINK export format</u>

## To load a PLINK export format

You can a retrieve a saved PLINK export format and run it on as-is needed based.

- 1. Open the Specify PLINK Export Settings dialog box. See To create a PLINK export
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Format dialog box opens.

Figure 10-8: Load Format dialog box



- 3. Open the folder that contains the PLINK export format that you are loading, select the format, and then click Load.
- 4. The Specify PLINK Export Settings dialog box is refreshed with the loaded format. You can modify the format as needed (for example, modify the information on the Markers tab), and export the format. See <a href="Creating and Running a PLINK Export"><u>Creating and Running a PLINK Export</u></a>

# Viewing and Rerunning a PLINK Export

You can view information about any PLINK exports that were previously run on the Analysis window, and you can rerun any PLINK export as necessary.

## To view and rerun a PLINK export

- 1. On the Analysis window, select the PLINK export that you want to view.
- 2. A single tab the Results tab opens in the bottom pane of the window.



You can also right-click on the tab and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 3. Optionally, to rerun the PLINK export, right-click on the selected export, and on the context menu that opens, click Open Export.
- 4. The Specify PLINK Export Settings dialog box opens. You can now modify any of the needed settings and click OK on the Settings tab to rerun the export. See Creating and Running a PLINK Export

# Deleting a PLINK Export

You can delete a PLINK export by doing one of the following:

- 1. Right-clicking on the export in the Analysis window, and on the context menu that opens, clicking Delete Export.
- 2. Selecting the export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Chapter 11 Helix Tree Exports

Progeny Lab provides all the functionality needed for creating Helix Tree exports and working with the results.

This chapter covers the following topics:

- **Creating and Running a Helix Tree Export**
- Saving and Loading a Helix Tree Export Format
- Viewing and Rerunning a Helix Tree Export
- **Deleting a Helix Tree Export**



To create and run Helix Tree exports, Helix Tree must be installed locally.

# Creating and Running a Helix Tree Export

Progeny Lab provides all the functionality needed for creating Helix Tree exports and working with the results. You must first create the export, and then you can run the export.

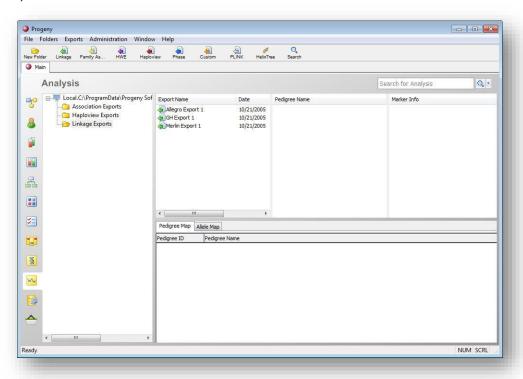


To create and run Helix Tree exports, Helix Tree must be installed locally.

## To create a Helix Tree export

1. On the main window navigation bar, click the Analysis button to open the Analysis window.

Figure 11-1: Analysis window



2. On the Analysis window, select the folder in which to the phase export is to be saved.

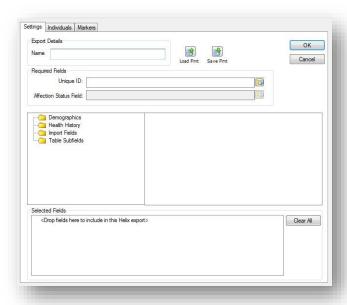


You can add a new folder if needed. Click the New Folder button on the window toolbar to open the New Folder dialog box, in which you can enter a name for a new Analysis folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the Phase HelixTree button. The Specify Helix Tree Settings dialog box opens. By default, the Settings tab is the active tab.
- 4. Specify the necessary export settings on all three tabs of the dialog box. See:
  - a. Specify Helix Tree Settings dialog box, Settings tab
  - b. Specify Helix Tree Settings dialog box, Individuals tab
  - c. Specify Helix Tree Export Settings dialog box, Markers tab
- 5. After you specify the necessary export settings on all three tabs, return to the Settings tab, and then click OK on the tab.
- 6. Continue To run a Helix Tree export

## Specify Helix Tree Settings dialog box, Settings tab

Figure 11-2: Specify Phase Export Settings dialog box, Settings tab



1. Specify the name for the export and the required fields for the export.

Option	Description	
Export Details		
Name	Name or description of the export.	
Required Fields		
Unique ID	Click the Fields button to the right of the Unique ID field to open the Select Field dialog box and select the field that represents the unique ID for everyone that is to be included in the export.	
Affection Status Field	Click the Fields button to the right of the Affection Status Field to open the Select Field dialog box and select the field that represents the affected status for everyone that is to be included in the export.	
	Note: This is appearing on the dialog box but is not available.	

- 2. Select the fields that are to be included in the export by doing the following:
  - a. In the Folders (middle left) pane of the dialog box, select the folder that contains the individual data fields that are to be included in the export.
  - b. In the Fields pane (middle right) pane of the dialog, select the fields (CTRL-click to select multiple fields) that are to be included in the export.
  - c. Drag the selected fields to the Selected fields (bottom) pane of the dialog box.

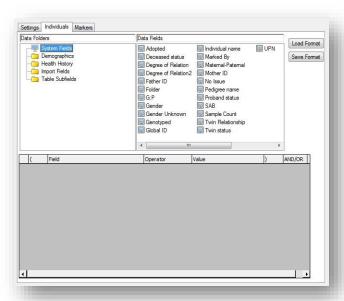


At least two fields must be selected to carry out a Helix Tree export.

3. Continue specifying any other Helix Tree export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a Helix Tree export</u>

## Specify Helix Tree Settings dialog box, Individuals tab

Figure 11-3: Specify Helix Tree Settings dialog box, Individuals tab



1. Define a query to select those individuals that are to be included in your Helix Tree export.

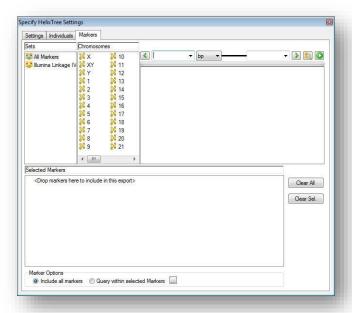


For detailed information about defining a query, see <u>Database Queries and</u> Searches

2. Continue specifying any other Helix Tree export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a Helix Tree export</u>

#### Specify Helix Tree Export Settings dialog box, Markers tab

Figure 11-4: Specify Helix Tree Settings dialog box, Markers tab



#### 1. Do one of the following:

- a. To include an entire marker set in the analysis, drag the marker set from the Sets (top right) pane of the dialog box to the Selected Markers (bottom) pane.
- b. To include all the markers for a specific chromosome in the analysis, drag the chromosome from the Chromosomes (top middle) pane of the dialog box to the Selected Markers (bottom) pane.
- c. To include only specific markers in the analysis, select the marker (CTRL-click to select multiple markers) from the top right pane to the Selected Markers (bottom) pane.



To search for a specific marker or markers, you can use the marker search functions. See Searching for a Marker

- 2. Select one of the following:
  - a. To include all the markers in the Selected Markers pane in the export, select Include all markers.
  - b. To include only those markers in the Selected Markers pane in the export that meet specific criteria, select Query within selected markers, and then click the Query button to open the Query dialog box and define a new query, or load a saved query format.

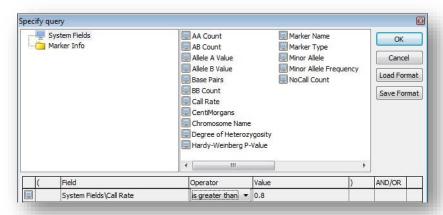


For detailed information about defining a query, see <u>Database Queries and</u> Searches



If you specify a query, then you can carry out additional marker filtering, and filter the selected SNPs by Hardy Weinberg p-value, Minor Allele Frequency, Degree of Heterozygosity, and/or Call Rates. For example, to include only those SNPs that have a call rate higher than 80%, you would select the Call Rate system field and enter > 0.8 as the operator and value. You can use this additional filtering criteria only if there is allele count data stored on a per SNP basis.

Figure 11-5: Additional marker filtering

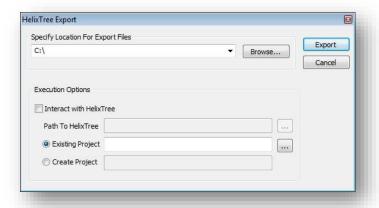


3. Continue specifying any other Helix Tree export settings as needed; otherwise, return to the Settings tab, click OK, and then continue <u>To run a Helix Tree export</u> below.

## To run a Helix Tree export

After you specify the necessary export settings on all three tabs, you must return to the Settings tab, and then click OK on the tab. The Helix Tree Export dialog box opens.

Figure 11-6: Helix Tree Export dialog box



1. Click Browse to open the Browse for Folder dialog box and browse to and select the folder in which the Helix Tree export files are to be stored.



If you have previously run Helix Tree exports, then any previously selected locations are available in a dropdown list in the Browse field. You can always select a value from this list.

- 2. Select the execution options:
  - a. Interact with Helix Tree Select this option to open Helix Tree after the files have been exported.
  - b. Path to Helix Tree Available only if you select Interact with Helix Tree. Click the Browse button to open the Browse for Folder dialog box and browse to and select the Helix Tree executable.
  - c. Select one of the following:
    - i. Existing Project—Select this option and then click the Browse button to open the Save As dialog box and select an existing Helix Tree projects to which to add these export files.
    - ii. Create Project—Select this option to create a new Helix Tree project.
- 3. Click Export. An Export to Helix Tree dialog box opens, indicating the status of the export.
- 4. When the export is complete, click Close to close the dialog box and return to the Analysis window.

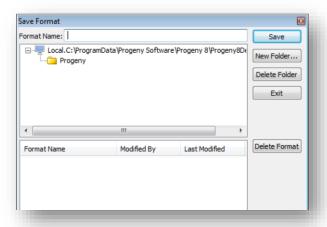
# Saving and Loading a Helix Tree Export Format

After you have set up a Helix Tree export format, you can save the format. You can then load this saved format and run it on as-needed basis.

## To save a Helix Tree export format

- 1. Set up the Helix Tree export format but do not run the format See Creating and Running a Helix Tree Export
- 2. Click the Save Format button Save Fmt on the Specify Helix Tree Settings dialog box.
- 3. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the export format. (This folder is in the database that you are currently logged in to and you cannot change this location.)

Figure 11-7: Save Format dialog box



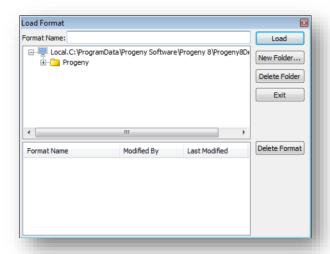
- 4. Enter a name for the export format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
  - c. Enter the name of the new folder in which to save to format.
  - d. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder. Click Save.
- 5. Click Save on the Save Format dialog box. You can then load this saved format and run it on as-needed basis. See To load a Helix Tree export format

## To load a Helix Tree export format

You can a retrieve a saved phase export format and run it on as-is needed based.

- 1. Open the Specify Helix Tree Settings dialog box. See To create a Helix Tree export
- 2. On the dialog box, click the Load Format button Load Format dialog box opens.

Figure 11-8: Load Format dialog box



- 3. Open the folder that contains the Helix Tree export format that you are loading, select the format, and then click Load.
- 4. The Specify Helix Tree Settings dialog box is refreshed with the loaded format. You can modify the format as needed (for example, modify the information on the Markers tab), and export the format. See <a href="Creating and Running a Helix Tree Export">Creating and Running a Helix Tree Export</a>

# Viewing and Rerunning a Helix Tree Export

You can view information about any Helix Tree exports that were previously run on the Analysis window, and you can rerun any phase export as necessary.

## To view and rerun a Helix Tree export

1. On the Analysis window, select the Helix Tree export that you want to view. A single tab—the Results tab—opens in the bottom pane of the window.



You can also right-click on the tab and open a context menu with options for copying the information on the tab, exporting the information on the tab to a text file, or printing the information on the tab.

- 2. Optionally, to rerun the Helix Tree export, right-click on the selected export, and on the context menu that opens, click Open Export.
  - a. The Specify Helix Tree Settings dialog box opens. You can now modify any of the needed settings (for example, modify information on the Markers tab) and click OK on the Settings tab to rerun the export. See Creating and Running a Helix Tree Export

# **Deleting a Helix Tree Export**

- 1. You can delete a phase export by doing one of the following:
  - a. Right-clicking on the export in the Analysis window, and on the context menu that opens, clicking Delete Export.
  - b. Selecting the export in the Analysis window, and then clicking the [Delete] key on your keyboard.

# Section 4 – Progeny LIMS

#### This appendix covers the following topics:

- Progeny LIMS Terms, Conventions and Components
- Managing Containers
- Managing Reactions
- Managing Actions
- Managing Samples
- Managing Workflows

# Chapter 1 – Progeny LIMS Terms, Conventions and Components

Progeny LIMS is software that is designed for managing any type of sample and associated data in a fully customizable multi-level inventory system with the freedom to create any laboratory workflow. Progeny LIMS uses conventions in both its terminology and its output to ensure consistency and accuracy when you are manipulating and analyzing sample data.

This chapter covers the following topics:

- Progeny LIMS Terms and Conventions
- Progeny LIMS Components

# **Progeny LIMS Terms and Conventions**

Progeny LIMS is software that is designed for managing any type of sample and associated data in a fully customizable multi-level inventory system with the freedom to create any laboratory workflow. Progeny LIMS uses conventions in both its terminology and its output to ensure consistency and accuracy when you are manipulating and analyzing sample data.

- 1. Sample A sample is a part that is representative of a whole or a set of elements that is drawn from and analyzed to estimate the characteristics of a population. A sample can be an unassociated sample, or it can be associated with an individual. The following icon indicates a sample:
- 2. Container A container is any receptacle in which a sample is held. A container can be as broad as a freezer and as a granular as a plate, for example, Freezer > Racks > Boxes > Plates. The following icon indicates a container:

# **Progeny LIMS Components**

To use all the features that are available to you in Progeny LIMS efficiently and effectively, you must understand the function/purpose of the following four critical components and their relationship with each other.

- 1. Sample database fields Sample database fields are stored at the sample record level. Sample database fields house the data that is entered for a sample in a Progeny database. Sample Name, Sample Type, Sample Status, and so on are examples of sample database fields. It is critical that you have defined all the necessary sample database fields in a Progeny database before you can work with any of the other three LIMS components. See Progeny Database Design.
- 2. Reactions A reaction is a system or user-defined trigger that is used to update sample records. Reactions can carry out a wide range of tasks, including, but not limited to, updating date sample data fields with set values or prompt users for input values, creating new sample and aliquot records, updating container locations, sending print commands to barcode label printers, assigning samples to individuals in the database and moving samples from one workflow to another. See Managing Reactions
- 3. Actions An action is a series of reactions. An action can be carried out on a single sample, a batch of samples or a container of samples, including plates. All the reactions that are contained in an action must be carried out successfully for the action to be completed. For example, the action "Update Sample Information" can consist of the following three reactions — "Update Sample Concentration", "Update Sample Status", and "Update Sample Type". To complete the "Update Sample" action, a user must enter the concentration for the sample (for example, 10 mg/dl), a sample type (for example, PreDosage or Post-Dosage), and a sample status (for example, Thawed or Frozen). See Managing Actions
- 4. Workflow A workflow, (also referred to as a sample workflow) is a collection of actions that have been arranged into a step-by-step procedure. A workflow is made up of multiple stages, with each stage associated with exactly one action. After a sample is placed into a workflow, the movement of the sample from one stage to the next can be tracked and recorded until the sample is ultimately exhausted and removed from the workflow. See Managing Workflows

# Chapter 2 - Managing Containers

A container is any receptacle in which a sample is held. A container can be as broad as a freezer and as a granular as a plate, for example, Freezer > Racks > Boxes > Plates. Managing containers consists of adding new containers, modifying and deleting containers, updating sample positions in containers, and importing and exporting container information.

This chapter covers the following topics:

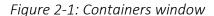
- Adding a New Container
- Modifying and Deleting a Container
- Manually Updating Sample Well Positions in a Plate or a Box
- Exporting Container Information
- Saving and Loading a Container Export File Format

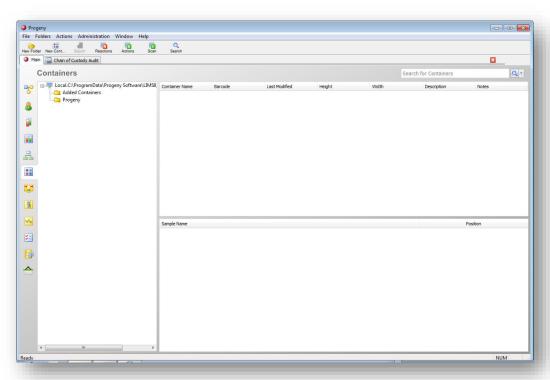
# Adding a New Container

A container is any receptacle in which a sample is held. A container can be as broad as a freezer and as a granular as a plate, for example, Freezer > Racks > Boxes > Plates. You can add the following containers in Progeny — freezer, shelf, box, refrigerator, rack, or plate. Adding a new container consists of naming the container and supplying a barcode and the dimensions for the container. In addition, if you are the Progeny administrator, you can set the security for the container. You can add a container as a standalone container, or you can add a container as a child container. For example, you can create a rack, and then from within the rack (the parent container), you can add a box or a plate (the child container).

#### To add a new container as a standalone container

1. On the Progeny main window navigation bar, click the "Containers" button to open the "Containers" window.





2. Select the folder to which the new container is being added.



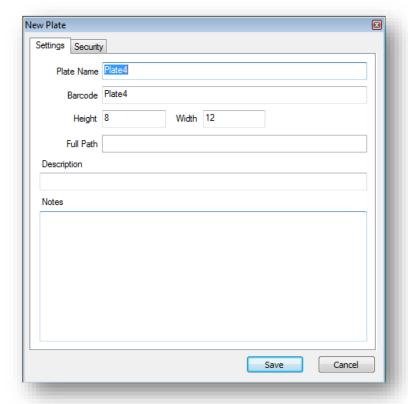
You can add a new folder if needed. Click the "New Folder" button on the window toolbar to open the "New Folder" dialog box, in which you can enter a name for a new Container folder and select its location (at the root level or as a sub-folder of another folder.)

- 3. On the window toolbar, click the "New Container" button to open a dropdown list of the containers that you can add, and then select the container that you are adding. The New Container dialog box opens.
- 4. Enter the information for the new container.



The following dialog box is that for a New Plate; however, for any new container, the New Container dialog box contains the same two tabs—a Settings tab for entering the container name, the container barcode, the container dimensions, and an optional description and/or notes about the container, and a Security tab for setting security for the container.

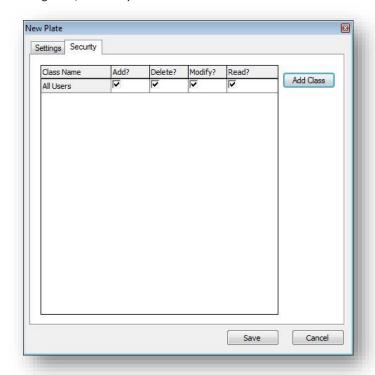
Figure 2-2: New Plate dialog box



Option	Description	
Container Name	The default name is ContainerTypeNumber, where ContainerType is Plate, Shelf, and so on, and Number is auto-incremented. For example, the first time that you add a new plate, the Plate Name is set to Plate1. If you use the default name for your containers, then the second time you add a plate, the default name is Plate2, and so on. You can modify the name to accommodate your own naming schemes.	
Barcode	The default barcode is the default Container Name. You can use this value, or you can modify it as needed.	
Height/Width	The dimensions for the container. Standard dimensions are used for the default values, but you can modify them as needed.	
Full Path	The full path to the container location for a shelf, box, rack or plate. For example, for a plate, the path could be -80Freezer\80_shelf\Rack1.	
	Note: If you add a shelf, box, rack, or plate as a child container, you do not have to enter this value manually, Instead, this field is automatically populated with the correct path and you cannot change this value. See <a href="To add a child container">To add a child container</a>	
Description/Notes	Optional fields	

5. Open the Security tab and set the security for the container.

Figure 2-3: New Container dialog box, Security tab



- 6. Do one or more of the following:
  - a. Modify the security for All Users.
  - b. Click Add Class to open the New User Class dialog box to create a new user class, and then set the security for the class.

Figure 2-4: New User Class dialog box

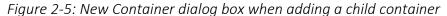


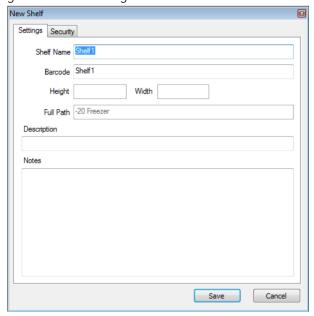
7. Click Save. The New Container dialog box closes. The new container is added to the Progeny database.

#### To add a child container

When you add a child container, the full path to the container is automatically populated, which provides an error free way of locating containers. You can add the following child containers:

- You can add a shelf to a freezer or refrigerator.
- You can add a box or a rack to a shelf.
- You can add a box or a plate to a rack.
- 1. Add the parent container. For example, if you want to add a shelf to a freezer, add the freezer. See <u>To add a new container</u> as a standalone container
- 2. Right-click on the parent container in the Containers window, and on the context menu that opens, select the child container that you are adding. For example, if you are adding a shelf to a freezer, then right-click on the freezer, and then click Add Shelf.
- 3. The New Container dialog box opens. The Full Path field is automatically populated for the child container and you cannot edit this value.





4. Enter the information for the child container as you would for any other container, and then click Save.

# Modifying and Deleting a Container

Any user can modify any of the non-security settings for a container. If you are the Progeny administrator, you can also modify the security settings. You can delete a container only if the container does not have any samples in it. If the container has samples, then you must first remove the samples from the container.

## To modify a container

- 1. On the Progeny main window navigation bar, click the Containers button to open the Containers window.
- 2. Open the data folder that contains the container that you are modifying.
- 3. Right-click on the container that you are modifying, and on the context menu that opens, click Edit. The Edit Container dialog box opens.
- 4. Edit the information for the container as needed, and then click Save.

Option	Description	
Container Name	The default name is ContainerTypeNumber, where ContainerType is Plate, Shelf, and so on, and Number is auto-incremented. For example, the first time that you add a new plate, the Plate Name is set to Plate1. If you use the default name for your containers, then the second time you add a plate, the default name is Plate2, and so on. You can modify the name to accommodate your own naming schemes.	
Barcode	The default barcode is the default Container Name. You can use this value or modify as needed.	
Height/Width	The dimensions for the container. Standard dimensions are used for the default values, but you can modify them as needed.	
Full Path	The full path to the container location for a shelf, box, rack or plate. For example, for a plate, the path could be -80Freezer\80_shelf\Rack1.	
	Note: If this container has been added as a child container, for example, the container is a rack that was added as a child container to a shelf, then this field is not editable.	
Description/Notes	Optional fields.	

#### To delete a container

- 1. On the Progeny main window navigation bar, click the Containers button it to open the Containers window.
- 2. Open the data folder that contains the container that you are deleting.
- 3. Right-click on the container that you are modifying, and on the context menu that opens, click Delete.



The Delete option is available only if there are no samples in the container. To delete a container with samples, you must first remove the samples from the container by running an action that contains the system reaction "Remove from Container." This reaction removes all samples from the container without deleting the sample records from the database. See <u>Carrying out Actions on a Sample in</u> the Scan Window

# Manually Updating Sample Well Positions in a Plate or a Box

After you have added a plate or box, you can manually update the positions of the samples in the plate or box. You can manually update the well positions by dragging samples to their correct positions, or you can simply enter the appropriate sample number for a well position.

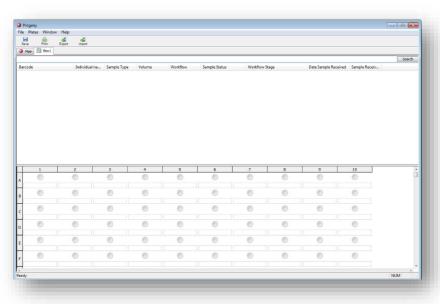


You can also use actions to automatically update sample positions in a container or remove a sample from a container. See <u>Managing Actions</u>

## To drag samples to their well positions in a plate or a box

- 1. On the Progeny main window navigation bar, click the Containers button to open the Containers window.
- 2. Open the data folder that contains the container for which you are modifying the well positions.
- 3. Double-click on the container for which you are modifying the well positions. The Container Viewer opens, and a search field is displayed at the top of the viewer.

Figure 2-6: Container Viewer

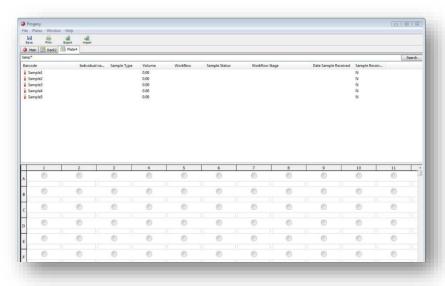


4. In the Search field, enter the criteria to search for the appropriate samples, and then click Search. The samples that meet the search criteria are displayed in the upper pane of the Container Viewer.



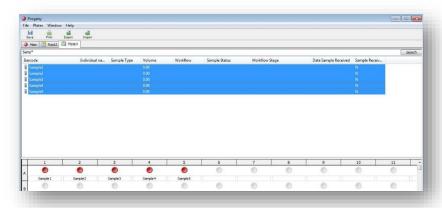
You search is limited to the exact order of the characters in the string and you can use the asterisk (\*) as a wildcard to extend the search. For example, Blood\* will return Blood1, Blood2, Blood\_1, Blood\_2, and so on.

Figure 2-7: Container Viewer with samples retrieved by a search



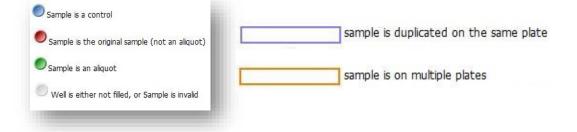
- 5. Select a sample (CTRL-click to select multiple samples), and then drag the selected samples to the appropriate position on the plate or box.
  - a. If you are dragging multiple samples, then the first selected sample is placed in the well to which the samples were dragged. The remaining samples are placed from left to right in the remaining available wells in the row, and then if applicable, moving down one row at a time, from left to right in the rows below.

Figure 2-8: Samples positioned in plate or box



The well colors indicate the type of sample that is placed in a well and the colors around the Sample Name field indicate whether a sample is duplicated on the sample plate or box or in multiple plates or boxes.

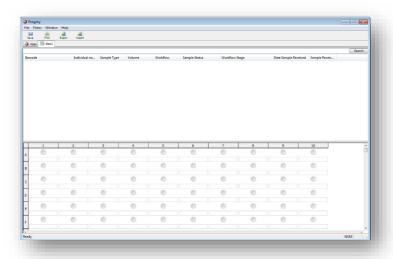
Figure 2-9: Color coding for samples in wells



## To manually enter positions for a sample in a plate or a box

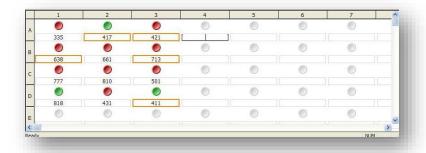
- 1. On the Progeny main window navigation bar, click the Containers button to open the Containers window.
- 2. Open the data folder that contains the container for which you are modifying the well positions.
- 3. Double-click on the container for which you are modifying the well positions. The Container Viewer opens.

Figure 2-10: Container Viewer



- 4. Select the text field below a well position, and then enter the sample number for the position.
  - a. If the sample has not yet been added to the Progeny database, then the well color does not change.

Figure 2-11: Manually entering samples in wells



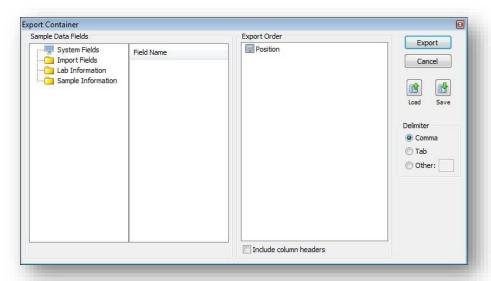
# **Exporting Container Information**

You can export container information to a text file.

## To export container information

- 1. On the Progeny main window navigation bar, click the Containers button to open the Containers window.
- 2. Open the data folder that contains the container for which you are exporting the information.
- 3. Select the container for which you are exporting the information, and on the Container window toolbar, click the Export button. The Export Containers dialog box opens.

Figure 2-12: Export Containers dialog box



- 4. In the Sample Data Fields (left) pane of the dialog box, open the data folder that contains the fields that you are exporting. The fields are displayed in the Field Name (middle) pane dialog box.
- 5. Drag the fields that are being exported to the Export Order (right) pane of the dialog box.



The order in which you drag the fields is the order in which the fields are exported in the text file. You can drag the fields in the Export Order pane to reorder them. To remove a field from the export, right-click on the field and click Remove.

- 6. Optionally, to include column headings in the export, select Include column headers.
- 7. Specify the delimiter for the export file. If the delimiter is not a tab or a comma, then you must select Other, and then enter the delimiter.
- 8. Click Export. The Save as Type field is automatically populated with the file type of text (.txt) and you cannot change this value.
- 9. Browse to the location in which the export file is to be saved, and then in the File name field, enter a name for the text file then click Save. A message opens indicating that the export was successful.
- 10. Click OK to close the message. The Export Container dialog box remains open.

# Saving and Loading a Container Export File Format

After you have set up a container export format, you can save the format. You can then load this saved format and run it on as-needed basis.

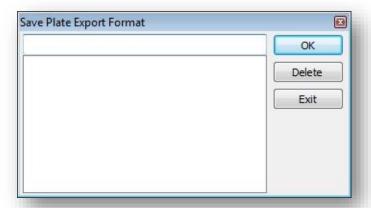
#### To save a container export format

- 1. Set up the container export format but do not run the format See Exporting Container Information
- 2. Click the Save Format Save Fmt button on the Export Container dialog box. The Save Plate Export Format dialog box opens.



Although the dialog box is named Save Plate Export Format, you can use this dialog box to save the export format for any container—freezer, shelf, box, and so on.

Figure 2-13: Save Plate Export Format dialog box



3. Enter a name for the container export format and click Save. You can then load this saved format and run it on as-needed basis. See <u>To load a container export format</u>

## To load a container export format

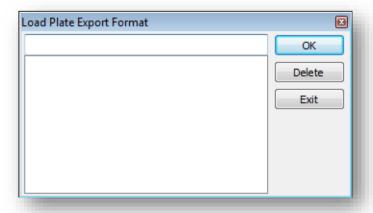
You can a retrieve a saved container export format and run it on as-is needed based.

- 1. Open the Export Container dialog box.
- 2. On the dialog box, click the Load Format Load Fmt button. The Load Plate Export Format dialog box opens.



Although the dialog box is named Load Plate Export Format, you can use this dialog box to load an export format for any container – freezer, shelf, box, and so on.

Figure 2-14: Load Plate Export Format dialog box



3. Select the container export format that you are loading, and then click Load. The Export Container dialog box is refreshed with the loaded format. You can modify the format as needed, and then export the format. See <a href="Exporting Container Information">Exporting Container Information</a>

# Chapter 3 Managing Reactions

A reaction is a system or user-defined trigger that is used to update sample records. Managing reactions consists of adding, modifying, and deleting reactions.

This chapter covers the following topics:

- Adding a Reaction
- Editing a Reaction
- Deleting a Reaction

# Adding a Reaction

A reaction is a system or user-defined trigger that is used to update sample records. A reaction always results in the updating or modifying of a single database field. For example, if a reaction is named Update Sample Status, and the reaction type is Update Value for the Sample Status field, then a prompt that could open for the Sample Status field is "Please Enter a Sample Status." You can add a reaction to a Progeny database from the Inventory window, the Containers window, or the Workflows window.

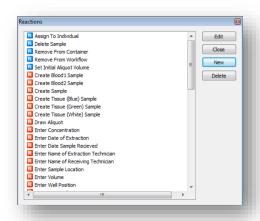


The following procedure describes how to add a new reaction for changing a field value and for defining sample barcode settings. You can also add reactions for creating and naming new samples and aliquots, but because creating and naming samples and aliquots are part of sample maintenance, these reactions are discussed in Managing Samples

#### To add a new reaction

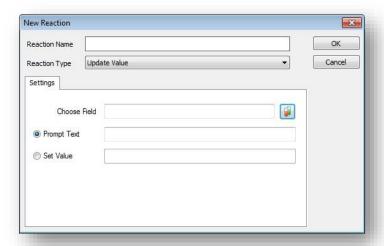
- 1. Open one of the following windows—Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Reactions Reactions button.
  - a. The Reactions dialog box opens. This dialog box lists all the reactions that are available in the Progeny database that you are currently logged in to. Reactions that are marked with a blue Reaction icon are system reactions that are always available in every Progeny database and are not editable. Reactions that are marked with a red Reaction icon are user-defined reactions and are editable.

Figure 3-1: Reactions dialog box



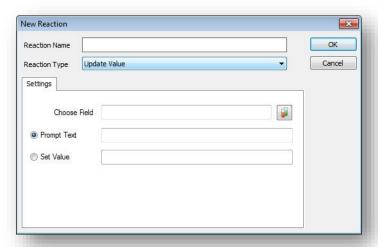
3. Click New. The New Reaction dialog box opens.

Figure 3-2: New Reaction dialog box



- 4. Enter the name for the new reaction and select the reaction type.
  - a. Update Value Override the current value in a data field. Go to Step 5.
  - b. Decrease Value Subtract an amount from current value in a data field. Go to Step 5.
  - c. Increase Value Add an amount to the current value in a data field. Go to Step 5.
  - d. Print Print the barcode label for a sample or send a print command to print a barcode label for a sample or batch of samples. Go to Step 6.
  - e. Create Sample See Adding a Create Sample Reaction
  - f. Create Aliquot See Adding a Create Aliquot Reaction
- 5. If you selected Update Value, Decrease Value, or Increase Value, then the New Reaction dialog box is refreshed with options for selecting the sample field on which to carry out the action, and whether the reaction is to be a prompt, or an automatic response.

Figure 3-3: New Reaction dialog box, Update Value selected



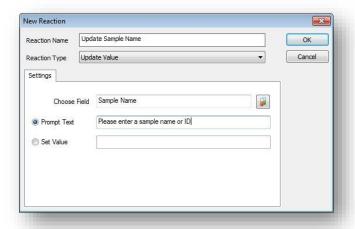
- 6. Do the following, and then click OK to add the new reaction.
  - a. Click the Sample button next to Choose Field to open the Select Field dialog box and select the field on which the action is to be carried out.
  - b. Select the reaction type Prompt or Set Value.



A Prompt is typically used for variables, such as "Please enter a Sample ID or Name." A Set Value is typically used for static values that required no manual intervention. Instead of a prompt appearing, the software automatically enters the value in the background, for example, "Mark Sample as Exhausted."

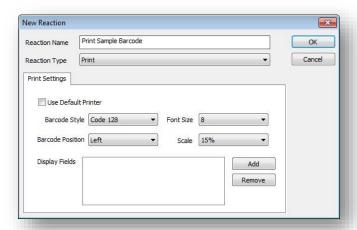
The figure below is an example of a reaction in which Update Value was selected for the Reaction Type. In this reaction, the value of the Sample Name field is to be updated, and the prompt that is given for the reaction is "Please Enter Sample Name or ID."

Figure 3-4: Example of an Update Value reaction



7. If you selected Print, then the New Reaction dialog box is refreshed with an option for printing the barcode from the default printer, as well as options for the barcode style, and the barcode position.

Figure 3-5: New Reaction dialog box, Print selected



- 8. Do the following, and then click OK to add the new reaction.
  - a. Optionally, select Use Default Printer. (If you do not select this option, then every time this reaction is run, a user is prompted to select a printer to use.)
  - b. Select the barcode style. Codes 128, 93, and 39 are standard one-dimensional barcoding languages. Data matrix is a two-dimensional barcoding language.
  - c. Select the barcode position (where the barcode is to be printed on the barcode label), font size, scale (the percentage of the barcode label that is to be used for printing the barcode), and display fields (the additional fields that are to be displayed on the barcode).

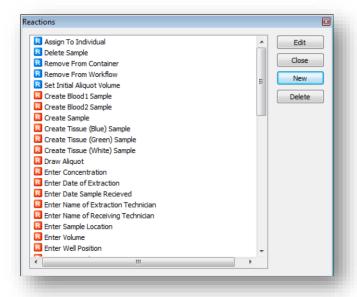
# **Editing a Reaction**

You can edit any user-defined reaction. You cannot edit the system reactions that are defined for every Progeny database.

#### To edit a reaction

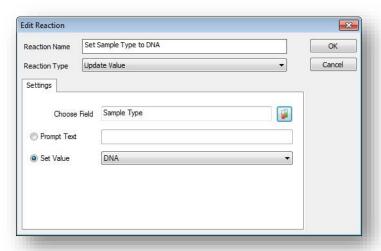
- 1. Open one of the following windows—Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Reactions Reactions button. The Reactions dialog box opens. This dialog box lists all the reactions that are available in the Progeny database that you are logged in to. Reactions that are marked with a blue Reaction icon are system reactions that are always available in every Progeny database and are not editable. Reactions that are marked with a red Reaction icon are user-defined reactions and are editable.

Figure 3-6: Reactions dialog box



3. Select the user-defined reaction that you are editing, and then click Edit. The Edit Reaction dialog box opens. The dialog box displays the reaction as it is currently defined.

Figure 3-7: Edit Reaction dialog box



4. Edit the reaction as needed, and then click OK to save the edited reaction.

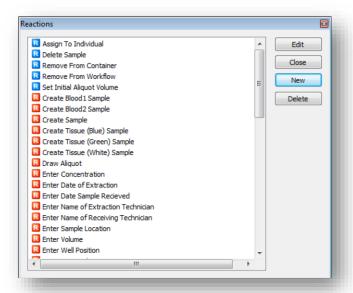
# **Deleting a Reaction**

You can delete any user-defined reaction. You cannot delete the system reactions that are defined for every Progeny database.

#### To delete a reaction

- 1. Open one of the following windows Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Reactions Reactions button. The Reactions dialog box opens. This dialog box lists all the reactions that are available in the Progeny database that you are logged in to. Reactions that are marked with a blue Reaction icon are system reactions that are always available in every Progeny database and are not editable. Reactions that are marked with a red Reaction icon are user-defined reactions and are editable.

Figure 3-8: Reactions dialog box



- 3. Select the user-defined reaction that you are deleting (CTRL-click to select multiple reactions), and then click Delete. A message opens asking you if you are sure that you want to delete the selected reactions.
- 4. Click Yes. The message closes, and the selected reactions are deleted.

# Chapter 4 Managing Actions

An action is a series of reactions. An action can be carried out on a single sample, a batch of samples, a container of samples, or a plate. Managing reactions consists of adding, modifying, and deleting actions.

This chapter covers the following topics:

- Adding an Action
- Setting an Action as a Default Action
- Specifying Action Barcode Settings
- <u>Setting Security for an Action</u>
- Editing an Action
- Deleting an Action

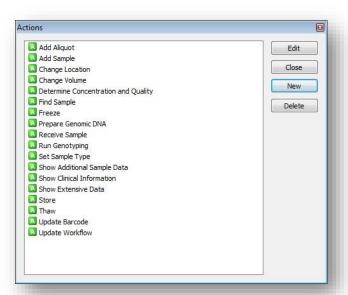
## Adding an Action

An action is a series of reactions. An action can be carried out on a single sample, a batch of samples, a container of samples, or a plate. You can add an action to a Progeny database from the Inventory window, the Containers window, or the Workflows window. When you add an action, you can add it "from scratch," or you can copy an existing action and modify it as necessary to make a new action.

#### To add a new action

- 1. Open one of the following windows—Inventory, Containers, or Workflow.
- 2. On the window toolbar, click the Actions Actions button. The Actions dialog box opens. This dialog box lists all the actions that have been defined for the Progeny database that you are logged in to.

Figure 4-1: Actions dialog box

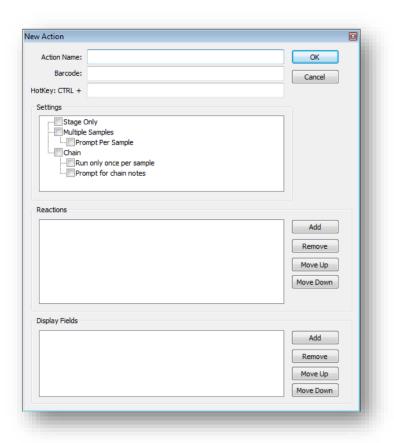




The remainder of this procedure describes how to add a new action "from scratch." To add a new action by copying an existing action, select the action that is to be copied, click Copy, and then continue to Step 4.

3. Click New. The New Action dialog box opens.

Figure 4-2: New Action dialog box



4. Enter the information for the new action.

Option	Description
Name	The name of the action.
Barcode	The barcode that is associated with the action. When this barcode is scanned into Progeny or manually entered in the Scan window, the action is initiated.  See <u>Carrying out Actions on a Sample in the Scan Window</u>
Burcouc	Note: You set the barcoding language, position, scale, and font for action barcodes in the Action Barcode Settings dialog box. See <a href="Specifying Action Barcode Settings">Specifying Action Barcode Settings</a>
Hotkey: CTRL +	The keystroke combination that initiates a selected action in the Scan window. See <u>Carrying out Actions on a Sample in the Scan Window</u>
	Select this option if the action is to be carried out only from within a workflow (a stage action).
Stage Only	Note: If the action is carried out outside of a workflow, then it is referred to as an open action.
Multiple Samples	Select this option if the action can be carried out on multiple samples at the same time. This is known as batch operation of samples.
Prompt Per Sample	Applicable only for Multiple Samples. A prompt opens for each selected sample when the action is initiated for the sample.
Chain	Select this option to log the action carried out on the sample into the Chain of Custody for the sample. The action is logged with the name of the user who carried out the action on the sample along with a date and timestamp.
Run only once per sample	Applicable only for Chain. Select this option to allow the indicated action to be carried out only once for any given sample if the action has already been logged into the Chain of Custody for the sample.
Prompt for chain notes	Applicable only for Chain. Select this option to allow the user to enter notes/comments about the action that is being logged into the Chain of Custody for the sample.
Prompt for number of samples to create	Displayed only if the first reaction in a series of reactions is of the type Create Sample and the reaction is set to Accept Default as the sample name. Select this option to open a prompt in which a user can specify the number of samples to create.
Prompt for number of aliquots to create	Displayed only if the first reaction in a series of reactions is of the type Create Aliquot and the reaction is set to Accept Default as the aliquot name. Select this option to open a prompt in which a user can specify the number of aliquots to create.

5. In the Reactions pane, click Add to open the Reactions dialog box and select the reactions (CTRL-click to select multiple reactions) that are to be contained in the action.



The order in which you add the reactions to the action is the order in which they are carried out. To change the order, select a reaction, and then click Move Up or Move Down as needed.

6. Optionally, in the Display Fields pane, click Add to open the Select Fields dialog box and select the fields that are to display information when the reaction is carried out.



The fields are for informational purposes only and are typically used to provide confirming or clarifying information for a reaction. For example, if you are entering the volume for an aliquot, and the aliquot naming scheme does not include the parent name, then you could select the Parent Name field for the reaction to confirm that you are entering the correct information for the correct aliquot.

7. Click OK to close the New Action dialog box and return to the Actions dialog box. The action is added to the Progeny database.

# Setting an Action as a Default Action

If needed, you can set an action to be your default action. A default action is initiated every time that you open the Scan window (see <u>Carrying out Actions on a Sample in the Scan Window</u>) from the Progeny main window toolbar. A default action is very similar to a hotkey for an action; however, you do not have to remember specific keystrokes for carrying out the action.

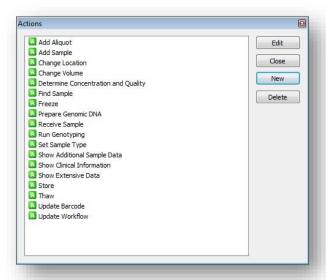


A default action is set on a per user basis. Every user can set up his/her own default action.

#### To set an action as a default action

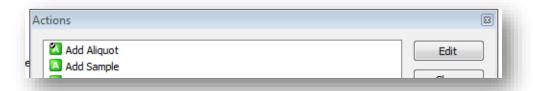
- 1. Open one of the following windows—Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Actions Actions button. The Actions dialog box opens. This dialog box lists all the actions that have been defined for the Progeny database that you are logged in to.

Figure 4-3: Actions dialog box



3. Right-click on the action that is to be your default action, and on the context menu that opens, click Set as Default Action. The action is marked with a black check mark in the Actions dialog box.

Figure 4-4: Default action indicated in the Actions dialog box





Conversely, to clear a default action, right-click on the action, and on the context menu that opens, click Remove as Default Action.

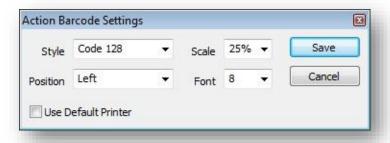
# **Specifying Action Barcode Settings**

When you create an action, you have the option of specifying a barcode for the action. When you scan this barcode into Progeny, or manually enter the barcode in the Scan window, the action is initiated. See <u>Carrying out Actions on a Sample in the Scan Window</u>. A very typical use for action barcodes is to create a "cheat sheet," which is essentially a piece of paper that contains labels, with each label having one action barcode printed on it. You can scan the barcodes on these labels to minimize your dependency on manually entering information to carry out a procedure in Progeny LIMS.

### To specify action barcode settings

- 1. Open one of the following windows Inventory, Containers, or Workflows.
- 2. On the window main menu, click Actions → Action Barcode Settings. The Action Barcode Settings dialog box opens.

Figure 4-5: Action Barcode Settings dialog box



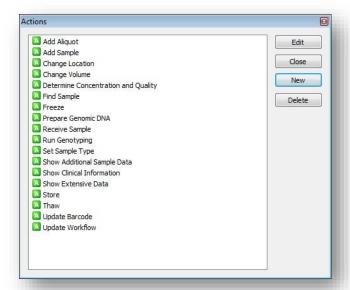
3. Specify the print settings for your action barcodes and indicate if are to use your system's default printer to print the barcodes.



The settings that you specify here are global settings and apply to all action barcodes. If you do not select Use Default Printer, then you when you print an action barcode, you are prompted to select a printer.

4. On the window toolbar, click the Actions Actions button. The Actions dialog box opens. This dialog box lists all the actions that have been defined for the Progeny database that you are logged in to.

Figure 4-6: Actions dialog box



5. For each action barcode that is to be printed on a label, right-click on the appropriate action then on the context menu that opens, click Print Barcode. The barcode is printed according to the settings that you specified in Step 3.



Make sure that you have the appropriate label sheet queued in the printer.

You can now paste all these printed labels on a sheet of paper and in the Scan window, use Progeny's barcode scanning function to scan a label to carry out an action. See <u>Carrying out Actions on a Sample in the Scan Window</u>. If you have a barcode label printed for every action that you must carry out, then you can carry out an entire procedure in LIMS without ever having to use a mouse and/or a keyboard.

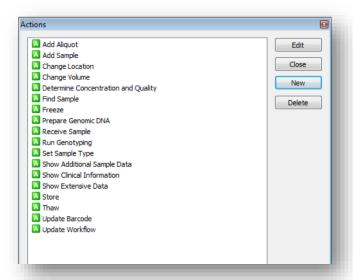
# Setting Security for an Action

If you are the Progeny administrator, then you can set security for actions.

### To set security for an action

- 1. Open one of the following windows—Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Actions button. The Actions dialog box opens. This dialog box lists all the actions that have been defined for the Progeny database that you are logged in to.

Figure 4-7: Actions dialog box



3. Right-click on an action, and on the context menu that opens, click Security Settings. The Action Security dialog box opens. This dialog box lists all the classes of users that have been defined for the database that you are logged in to.

Figure 4-8: Action Security dialog box



- 4. Modify the security for All Users or for the different classes of users.
  - a. Delete? The users can delete the action.
  - b. Modify? The users can modify the action.
  - c. Read? The users can carry out the action.
- 5. Click OK.

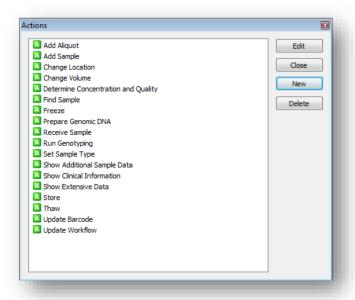
# **Editing an Action**

If your administrator has set the correct security, then you can edit an action. As the Progeny administrator, you can always edit any action.

### To edit an action

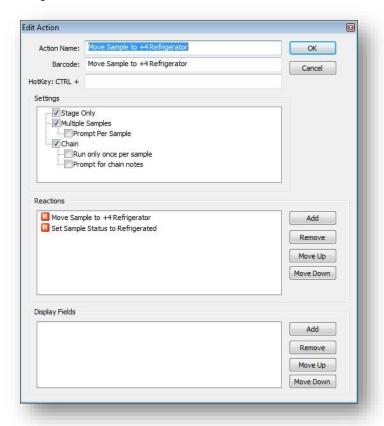
- 1. Open one of the following windows—Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Actions Actions button. The Actions dialog box opens. This dialog box lists all the actions that have been defined for the Progeny database that you are logged in to.

Figure 4-9: Actions dialog box



3. Select the action that you are editing, and then click Edit. The Edit Action dialog box opens. The dialog box displays the action as it is currently defined.

Figure 4-10: Edit Reaction dialog box



4. Modify the action as needed, and then click OK to save the modified action.

# **Deleting an Action**

If your administrator has set the correct security, then you can delete an action. As the Progeny administrator, you can always delete any action.

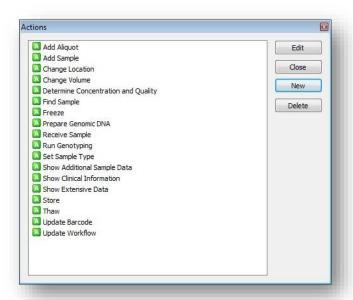


If you delete an action that is part of a workflow, then the action is also deleted from the workflow. You must edit the workflow to accommodate the gap in the workflow. See <u>Managing Workflows</u>

#### To edit an action

- 1. Open one of the following windows—Inventory, Containers, or Workflows.
- 2. On the window toolbar, click the Actions Actions button. The Actions dialog box opens. This dialog box lists all the actions that have been defined for the Progeny database that you are logged in to.

Figure 4-11: Actions dialog box



- 3. Select the action that you are deleting (CTRL-click to select multiple actions), and then click Delete. A message opens prompting you to delete the selected reactions.
- 4. Click Yes. The message closes and the selected reactions are deleted from the Progeny database.

# Chapter 5 - Managing Samples

A sample is a part that is representative of a whole or a set of elements that is drawn from and analyzed to estimate the characteristics of a population. A sample can be an unassociated sample, or it can be associated with an individual. Managing samples in Progeny LIMS consists of creating new samples, carrying out actions on samples, generating sample reports, assigning samples to workflows and moving a sample through a workflow, modifying samples, deleting samples, importing sample data. In Progeny LIMS, you accomplish all these tasks through use of actions.

This chapter covers the following topics:

- Adding a Create Sample Reaction
- Adding a Create Aliquot Reaction
- Carrying out Actions on a Sample in the Scan Window
- Creating a New Sample Datasheet
- **Generating a Chain of Custody Audit Report**



For detailed information about importing sample data, see Appendix B: Importing Clinical Data. For detailed information about assigning a sample to a workflow and moving a sample through a workflow, see

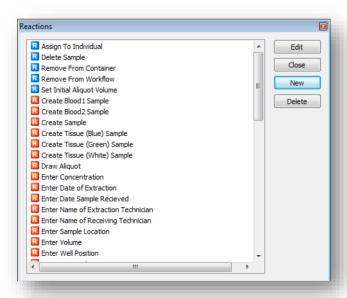
### Adding a Create Sample Reaction

A reaction is a system or user-defined trigger that is used to update sample records. A reaction always results in the updating or modifying of a single database field. In Progeny LIMS, you use reactions to create new samples. When you add a reaction for creating new samples, you must also define the naming schema for the samples. You can add a single reaction that simply sequentially numbers all samples as they are added to a Progeny database, or you can add multiple reactions with each reaction creating a different type of sample and each sample type having a unique naming schema. For example, you could add a reaction named Create Blood Samples with a naming schema that results in samples named Blood1, Blood2, Blood3, and so on and you could add a reaction named Create Tissue Samples with a naming schema that results in samples named Tissue1, Tissue2, Tissue3, and so on.

#### To add a create sample reaction

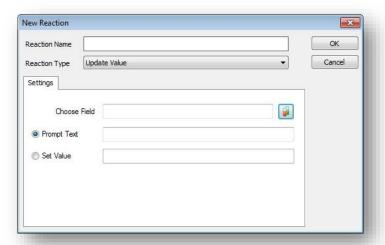
- 1. Open one of the following windows—Inventory, Containers, or Workflow.
- 2. On the window toolbar, click the Reactions Reactions button. The Reactions dialog box opens. This dialog box lists all the reactions that are available in the Progeny database that you are currently logged in to.

Figure 5-1: Reactions dialog box



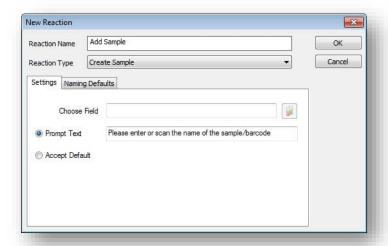
3. Click New. The New Reaction dialog box opens.

Figure 5-2: New Reaction dialog box



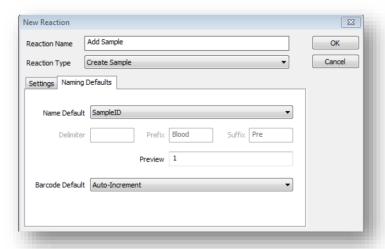
- 4. In the Reaction Name field, enter an appropriate name such as Add Sample or Create Sample.
- 5. On the Reaction Type dropdown list, select Create Sample. The New Reaction dialog box is refreshed with two tabs Settings and Naming Defaults. The Settings tab is the active tab.

Figure 5-3: New Reaction dialog box, Settings tab



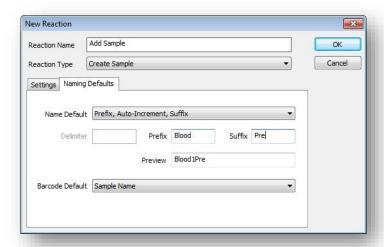
- 6. Do one of the following:
  - a. If the sample barcodes were not originated in Progeny (that is you are going to scan sample barcodes as the samples are received), then select Prompt Text, and in the Prompt Text field, enter the prompt that is given to a user for adding a new sample.
  - b. If the sample barcodes originated in Progeny (that is, the sample barcodes are already in the Progeny database), then select Accept Default.
- 7. Open the Naming Defaults tab, do one of the following to specify the naming scheme for your samples, and then click OK to create the reaction.
  - a. Leave the Name Default set to SampleID and Barcode Default to Auto-increment to increment every sample that is added to the database. The sample names are simply sequential numbers -1, 2, 3, and so on as shown in the Preview field.

Figure 5-4: New Reaction dialog box, Naming Defaults tab



b. Select Prefix, Auto-Increment, Suffix for the Name Default, and select Sample Name for Barcode Default to create a unique barcode naming convention that autoincrements every sample that is added to the database. The sample names are sequentially numbered and are uniquely identified using a prefix and/or suffix, for example, Blood1Pre, Blood2Pre, Blood3Pre, and so on as shown in the Preview field.

Figure 5-5: New Reaction dialog box, Naming Defaults tab



With this approach, you can create multiple reactions with each reaction creating a different type of sample and each sample type having a unique naming schema. For example, you could create a reaction named Create Blood Samples with a naming schema that results in samples named Blood1, Blood2, and so on, and you could create a reaction named Create Tissue Samples with a naming schema that results in samples named Tissue1, Tissue2, and so on.

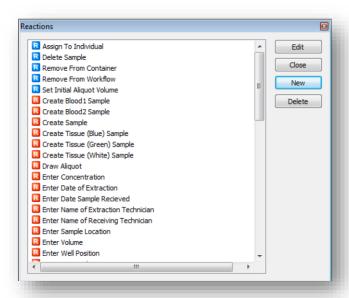
## Adding a Create Aliquot Reaction

A reaction is a system or user-defined trigger that is used to update sample records. A reaction always results in the updating or modifying of a single database field. In Progeny LIMS, you use reactions to create new aliquots. When you add a reaction for creating new aliquots, you must also define the naming schema for the aliquots. You can add a single reaction that simply sequentially numbers all aliquots as they are added to a Progeny database, or you can add multiple reactions with each reaction creating a different type of aliquot and each aliquot type having a unique naming schema. For example, you could add a reaction named Create Blood Aliquots with a naming schema that results in samples named Blood1, Blood2, Blood3, and so on and you could add a reaction named Create Serum Aliquots with a naming schema that results in samples named Serum1, Serum2, Serum3, and so on.

#### To add a create aliquot reaction

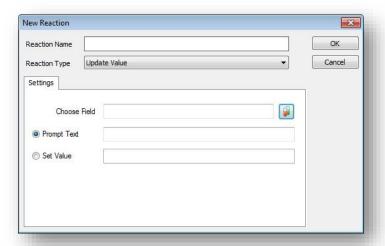
- 1. Open one of the following windows—Inventory, Containers, or Workflow.
- 2. On the window toolbar, click the Reactions Reactions button. The Reactions dialog box opens. This dialog box lists all the reactions that are available in the Progeny database that you are currently logged in to.

Figure 5-6: Reactions dialog box



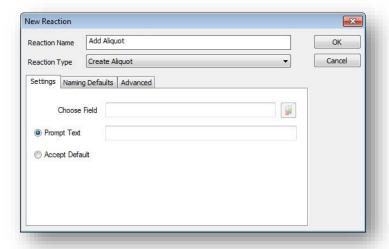
3. Click New. The New Reaction dialog box opens.

Figure 5-7: New Reaction dialog box



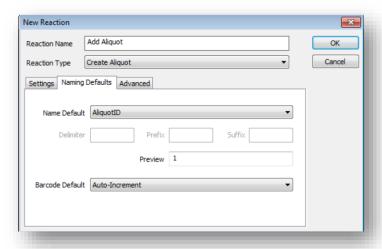
- 4. In the Reaction Name field, enter an appropriate name such as Add Aliquot or Create Aliquot.
- 5. On the Reaction Type dropdown list, select Create Aliquot. The New Reaction dialog box is refreshed with three tabs—Settings, Naming Defaults, and Advanced. The Settings tab is the active tab.

Figure 5-8: New Reaction dialog box, Settings tab



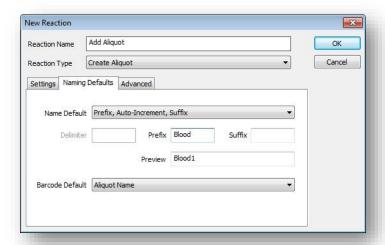
- 6. Do one of the following:
  - a. If the sample barcodes were not originated in Progeny (that is you are going to scan sample barcodes as the samples are received), then select Prompt Text, and in the Prompt Text field, enter the prompt that is given to a user for adding a new sample.
  - b. If the sample barcodes originated in Progeny (that is, the sample barcodes are already in the Progeny database), then select Accept Default.
- 7. Open the Naming Defaults tab, and then do one of the following to specify the naming scheme for your samples.
  - a. Leave the Name Default set to SampleID and Barcode Default to Auto-increment to increment every sample that is added to the database. The sample names are simply sequential numbers -1, 2, 3, and so on as shown in the Preview field.

Figure 5-9: New Reaction dialog box, Naming Defaults tab



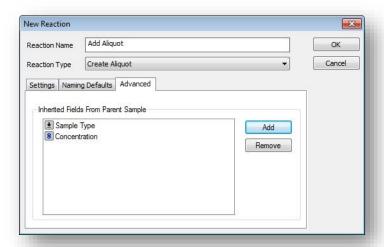
- b. Select Prefix, Auto-Increment, Suffix for the Name Default, and select Aliquot Name for Barcode Default to create a unique barcode naming convention that autoincrements every sample that is added to the database. The sample names are sequentially numbered and are uniquely identified using a prefix and/or suffix, for example, Blood1, Blood2, Blood3, and so on as shown in the Preview field.
- c. Select Parent Sample Name, Delimiter, Auto-Increment and select Aliquot Name for Barcode Default to create a unique barcode naming convention that autoincrements every sample that is added to the database. The sample names are sequentially numbered and are uniquely identified using the parent name and a delimiter, for example, SampleParentName\_1, SampleParent\_2, SampleName\_3, and so on as shown in the Preview field.

Figure 5-10: New Reaction dialog box, Naming Defaults tab



- d. With this approach, you can create multiple reactions with each reaction creating a different type of aliquot and each aliquot type having a unique naming schema. For example, you could create a reaction named Create Blood Aliquots with a naming schema that results in samples named Blood1, Blood2, Blood3, and so on and you could create a reaction named Create Serum Aliquots with a naming schema that results in samples named Serum1, Serum2, Serum3, and so on.
- 8. Open the Advanced tab, and then click Add to open the Select Fields dialog box to select the data fields that the aliquot is to inherit from the parent sample.

Figure 5-12: New Reaction dialog box, Advanced tab



9. Click OK to add the new reaction.

### Carrying out Actions on a Sample in the Scan Window

An action is a series of reactions. An action can be carried out on a single sample, a batch of samples, or a container of samples, including plates. All the reactions that are contained in an action must be carried out successfully for the action to be completed. In Progeny LIMS, you carry out actions on a sample, either standalone, or when it is included in workflow stages, in the Scan window. You can open the Scan window in one of three ways:

- By clicking the Scan button scan on the toolbar for the Samples, Inventory, Workflows, or Containers windows.
- By right-clicking on a selected sample, a batch of samples, or a container, including plates, and on the context menu that opens, clicking Actions, and then selecting an action.
- By selecting a sample, a batch of samples, or a container, including plates, and then entering the appropriate Hotkey combination (for example, CTRL + s) for an action.

The Scan window displays critical information about a sample, including its history, the sample or sample batch number, and the current sample action and reaction. The Scan window also contains options for canceling the current action, retaining the current action, and manually entering or scanning action barcodes. After your LIMS implementation is completely operational, you can limit your interaction with Progeny solely to this window.

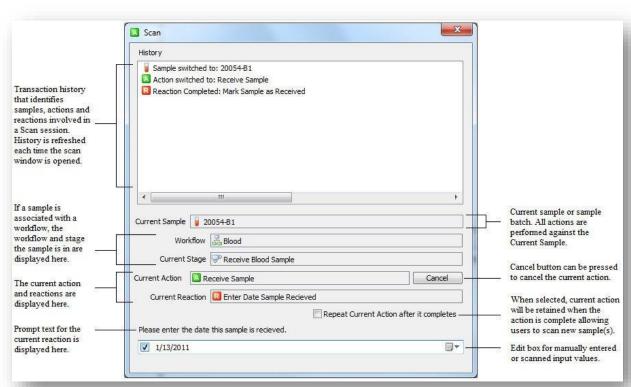
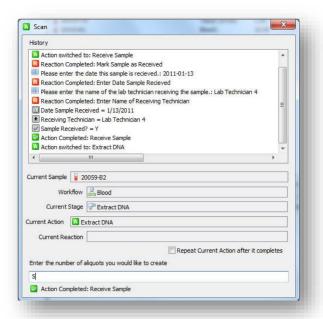
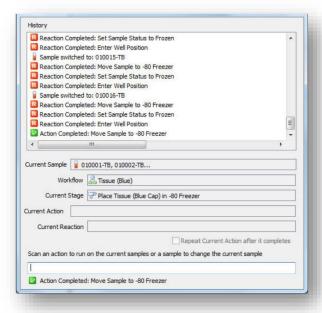


Figure 5-13: Scan window

Figure 5-14 below show actions being carried out on a single sample in the Scan window and for a batch of samples in the Scan window. Both windows show the history of an action being carried out from beginning to end. In addition, Figure 5-14 shows that multiple actions can be carried out for samples in a single Scan session. You do not have to open a Scan window for each action that you want to carry out on a sample.

Figure 5-14: Carrying out multiple actions for a single sample or multiple samples in the Scan window







For assistance with setting up Scan modes for your samples, contact support@progenygenetics.com

### Creating a New Sample Datasheet

You use a sample datasheet to manually enter and store data for a sample in a Progeny database. All the samples that are assigned to a sample data folder can use the same datasheet format, or each sample in the folder can use a unique datasheet format. You can create and format a sample datasheet at the time that you add a sample to a data folder, or later.

#### To create a sample datasheet

- 1. Create a sample data folder.
  - a. If all the samples that are assigned to the folder are to use the same sample datasheet format, make sure to select the Make this folder a template folder option; otherwise, clear it. See <u>Adding Sample</u> <u>Data Folders</u>
- 2. Do one of the following:
  - a. Create a new sample and add the sample to the data folder. Double-click on the sample in the folder to open a blank datasheet and format the datasheet as needed. If the Make this folder a template folder option has been selected for the data folder, then all the samples that you subsequently add to the folder will use the same datasheet format for entering and storing data; otherwise, you must format a datasheet for each sample that you add to the folder.
  - b. Add all the needed samples to the data folder. After you have added all the needed samples to the folder, double-click on any sample in the folder to open a blank datasheet and format the datasheet as needed. If the Make this folder a template folder option has been selected for the data folder, then all samples that are currently added to the folder or that you subsequently add to the folder will use this same datasheet format for entering and storing data; otherwise, you must format a datasheet for each sample in the folder.



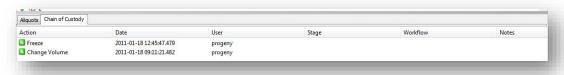
If data has been added to a sample through an action, and the database field that holds this data has been added to the sample datasheet (for example, Sample Type), then when you open the sample datasheet, this field is already populated with the data (for example, Frozen or Thawed).

3. See <u>Creating and Formatting Datasheets</u> for detailed information about creating and formatting a sample datasheet.

## Generating a Chain of Custody Audit Report

When you are creating an action, you have the option of including the action in the chain of custody. The chain of custody is an action audit trail. Any action that has been marked for inclusion in the chain of custody is timestamped with the time that the action was carried out, the name of the user (user name) who carried out the action, the action ID, and if the sample is included in a workflow, the workflow stage ID and the workflow ID. You can generate a chain of custody for every sample in a Progeny database. This information is displayed on the Chain of Custody tab on the Samples window and on the Inventory window.

Figure 5-15: Chain of Custody tab

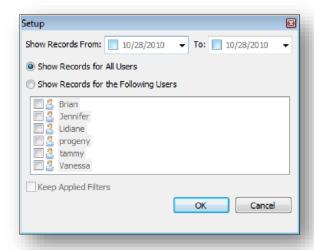


You can generate a Chain of Custody Audit report to filter the Chain of Custody records for every sample in a Progeny database.

#### To generate a Chain of Custody Audit report

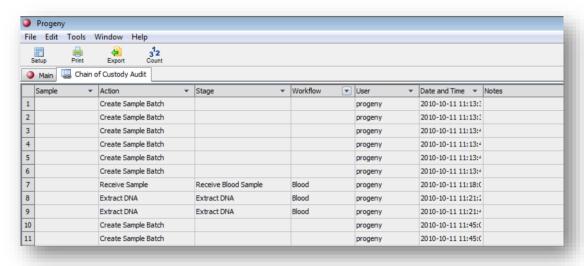
1. Open the Samples or Inventory window, and on the main menu, click Administration → Chain of Custody Audit. The Setup dialog box opens.

*Figure 5-16: Setup dialog box* 



- 2. Set the report filtering options:
  - a. Date The default value for both the From and To dates is the current day's date. Select a From date and/or a To date. Leave both date fields blank to retrieve all records in the database. Leave date field blank to have the results be open-ended. For example, if you enter a From date and leave the To date empty, all records that were generated between the From date and the current day's date are retrieved.
  - b. User The default value is to show the records for all users. You can select Show Records for the following users, and then select the specific users for whom to retrieve the records.
- 3. Click OK. The Chain of Custody Audit report is generated onscreen in a spreadsheet format.

Figure 5-17: Chain of Custody Audit report



- 4. Optionally, after you generate an Chain of Custody Audit report, you can do one or more of the following:
  - a. Sort and/or filter the audit data. See Sorting and Filtering Spreadsheet Data
  - b. Click the Setup button Setup to open the Audit setup dialog box, modify the report settings and generate the report again.
  - c. Click the Print button to print the Audit report.
  - d. Click the Export Export button to export the Audit report. See Exporting Spreadsheet Data
  - e. Click the Count button count the number of individuals, pedigrees, or samples (that is, the number of rows) in the spreadsheet. See Counting Spreadsheet Data

## Chapter 6 Managing Workflows

Workflow – A workflow, (also referred to as a sample workflow) is a collection of actions that have been arranged into a step-by-step procedure. A workflow is made up of multiple stages, with each stage associated with exactly one action. After a sample is placed into a workflow, the movement of the sample from one stage to the next can be tracked and recorded until the sample is ultimately exhausted and removed from the workflow. Managing a workflow includes creating, modifying, and deleting workflows, assigning a sample to a workflow, and moving a sample through a workflow.

This chapter covers the following topics:

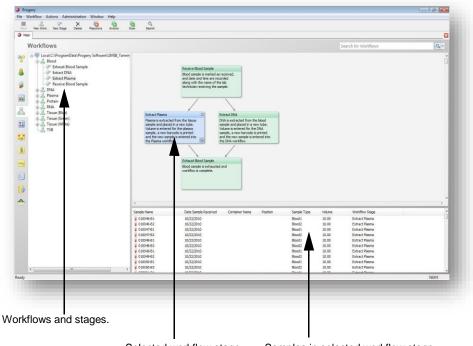
- Workflows Window
- Creating a New Workflow
- Setting Workflow Security
- Modifying and Deleting a Workflow Stages
- Deleting a Workflow
- Assigning a Sample to a Workflow
- Moving Samples through a Workflow

#### **Workflows Window**

The Workflow function in Progeny allows you to create a workflow that outlines the path that a sample must take as it is moved through a laboratory. You have full control to decide the stages the sample must pass through and the action to be carried out on sample at each stage of the workflow. The Workflows window displays information about every workflow that you have created in a Progeny database.

- The left pane in the Workflows window lists all the workflows that are available in a Progeny database. Select a workflow to view all the stages associated with a workflow.
- The top right pane is a graphical representation of a workflow, or a workflow drawing, with each stage identified in a green square. By default, the first time that you select a workflow in the left pane, all the samples that are assigned to a workflow are displayed in the bottom right pane of the window. To view only those samples in a specific stage, select the stage. The selected stage is highlighted in blue, and all the samples that are assigned to the stage are displayed in the bottom right pane of the window.

Figure 6-1: Workflows window



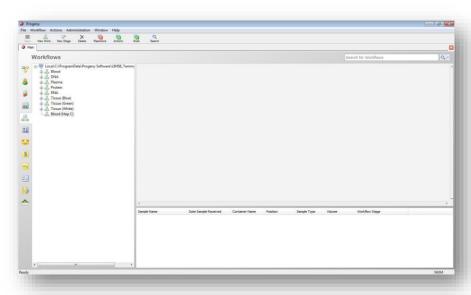
## Creating a New Workflow

Creating a new workflow consists of three steps—naming the workflow, adding stages to the workflow, and connecting the workflow stages.

#### To name a workflow

1. On the Progeny main window navigation bar, click the Workflows button to open the Workflows window.

Figure 6-2: Workflows window



2. On the window toolbar, click the New Workflow New Workflow button. The New Workflow dialog box opens.

Figure 6-3: New Workflow dialog box

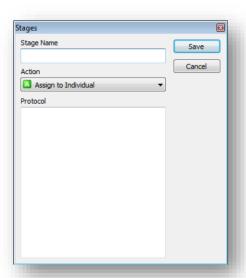


- 3. Enter a name for the new workflow, and then click OK. The New Workflow dialog box closes. The new workflow is displayed in the right pane of the Workflows window and by default, it is the selected workflow.
- 4. Continue To add stages to a workflow

#### To add stages to a workflow

- 1. If you have not already done so, open the Workflows window and select the workflow to which you are adding the stages.
- 2. On the window toolbar, click the New Stage New Stage button. The Stages dialog box opens.

Figure 6-4: Stages dialog box



- 3. Enter the name for the stage and on the Action dropdown list, select the action for the stage.
- 4. Optionally, in the Protocol field, enter a description about the stage.



The stage name and protocol are displayed on the workflow graphic in the right pane of the Workflows window.

- 5. Click Save. Repeat until you have added all the necessary stages for the workflow.
- 6. Continue To connect workflow stages

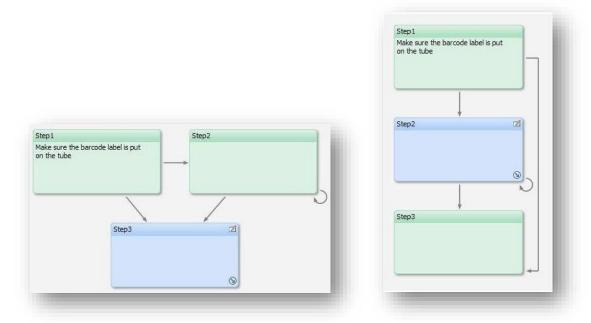
#### To connect workflow stages

- 1. If you have not already done so, open the Workflows window and select the workflow for which you are connecting the stages.
- 2. Connect or disconnect the stages as needed doing one or more of the following:
  - a. To connect one stage to another stage, click and drag the Arrow icon in the lower right corner. (Figure 6-5)
  - b. To indicate that a stage is to be repeated, click and drag the Arrow icon for the stage to the center of the stage graphic. The Arrow icon changes to a Repeat icon. (Figure 6-6)
  - c. To disconnect a stage, repeat the action that connected the two stages. For example, if you connected Stage 1 to Stage 2, and decide that you need to disconnect Stage 1 from Stage 2, simply repeat the action for connecting Stage 1 and Stage 2 and the stages are disconnected. To remove the Repeat indication from a stage, simply click and drag the Arrow icon for the stage to the center of the stage graphic icon again.



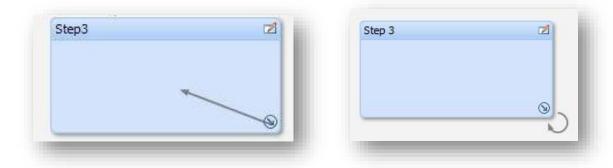
When you are connecting workflow stages, you can connect them horizontally, vertically, or use a combination of these. Figure 6-5 below illustrates this concept. Both workflows that are shown are identical.

Figure 6-5: Workflow orientation



- Step1 is the only starting point of the workflow. When a sample is assigned to the workflow, Step1 is the first Next Stage.
- Step1 has Next Stages of Step2 or Step3.
- Step2 has Next Stages of Step2 or Step3.
- Step3 is the conclusion of the workflow.

Figure 6-6: Indicating that the workflow stage is to be repeated



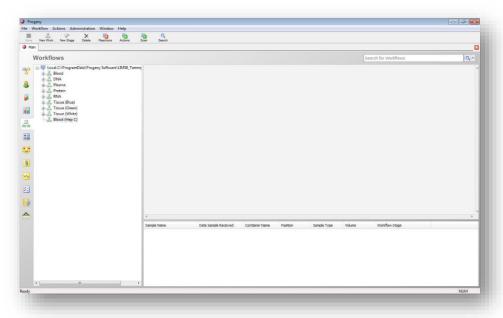
## **Setting Workflow Security**

As the Progeny administrator, you can set the security for a workflow.

#### To set security for a workflow

1. On the Progeny main window navigation bar, click the Workflows button to open the Workflows window.

Figure 6-7: Workflows window



2. Right-click on the workflow for which you are setting the security and click Security Settings. The Workflow Security dialog box opens. This dialog box lists all the classes of users that have been defined for the database that you are logged in to.

Figure 6-8: Workflow Security dialog box



- 3. Modify the security for All Users or for the different classes of users.
  - a. Delete? The users can delete the workflow and/or stages in the workflow.
  - b. Modify? The users can modify the workflow and/or stages in the workflow.
  - c. Read? The users can carry out the workflow.
- 4. Click OK.

### Modifying and Deleting a Workflow Stages

If your administrator has assigned you the correct security, you can modify workflow stages even if samples are currently assigned to the stage. As the Progeny administrator, you can always modify workflow stages even if samples are currently assigned to the stage. If your administrator has assigned you the correct security, you can delete a workflow stages if no samples are assigned to the stage. As the Progeny administrator, you can always delete a workflow stages if no samples are assigned to the stage. If samples are assigned to a stage, you must first move all the samples that are in the stage onto the next stage.

#### To modify a workflow stage

- 1. On the Progeny main window navigation bar, click the Workflows button to open the Workflows window.
- 2. Select the workflow for which you are modifying the stage. The workflow drawing is displayed in the top right pane of the window.
- 3. Click the Edit button in the upper right-hand corner of the stage that you are modifying. The Stage dialog box opens.
- 4. Modify the information as needed for the stage, and then click Save.

#### To delete a stage

- 1. On the Progeny main window navigation bar, click the Workflows button to open the Workflows window.
- 2. Select the workflow for which you are deleting the stage. The workflow drawing is displayed in the top right pane of the window.
- 3. Select the stage that you are deleting either in the workflow drawing, or in the right pane of the Workflows window, and then click the Delete button pelete on the window toolbar. A message opens asking you if you are sure that you want to delete the stage.
- 4. Click Yes. The messages close and the stage is deleted.

## Deleting a Workflow

If your administrator has assigned you the correct security, you can delete a workflow only if there are no samples assigned to the workflow, or if no samples are currently being moved through the workflow. As the Progeny administrator, you can always delete a workflow only if there are no samples assigned to the workflow, or if no samples are currently being moved through the workflow. If samples are assigned to the workflow, you must first remove the samples from the workflow, or move the samples through the workflow to complete the workflow.

#### To delete a workflow

- 1. On the Progeny main window navigation bar, click the Workflows button to open the Workflows window.
- 2. Select the workflow that you are deleting.
- 3. On the window toolbar, click the Delete button Delete . A message opens asking you if you are sure that you want to delete the workflow.
- 4. Click Yes. The messages close and the workflow is deleted.

## Assigning a Sample to a Workflow

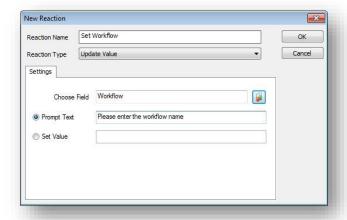
After you have created a workflow, you can assign the needed samples to the workflow. To assign a sample to a workflow you must first create an Update Workflow reaction. After you create this action, you can use it repeatedly to assign any number of samples to any workflow.

#### To assign a sample to a workflow

1. Add a reaction named Set Workflow. See Adding a Reaction

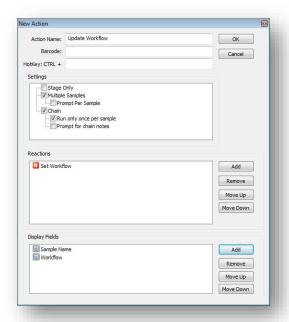


Figure 6-9: Adding the Set Workflow reaction



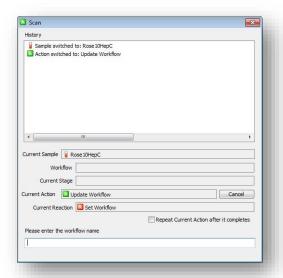
2. Add an action named Update Workflow that includes the Set Workflow reaction. See Adding an Action

Figure 6-10: Adding the Update Workflow action



- 3. Open the Inventory or Samples window and locate the sample that you are adding to the workflow.
- 4. Right-click on the sample, and on the context menu that opens, click Actions → Update Workflow. The Scan dialog box opens. The cursor is in the field that prompts you to enter the workflow name.

Figure 6-11: Scan dialog box for adding a sample to a workflow



- 5. Enter the name of the workflow, and then press [Enter] on your keyboard.
- 6. The Scan dialog box is refreshed with the message Action Completed: Update Workflow. The sample can now be moved through the stages of the workflow. See <u>Moving Samples through a Workflow</u>



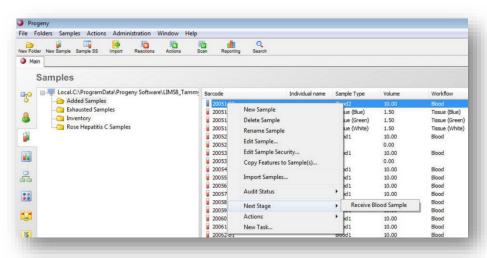
You must enter the name exactly as it is shown on the Workflows window (including capitalization and spacing) or an error message opens indicating that the workflow does not exist.

## Moving Samples through a Workflow

Three options are available to you for moving a sample or a batch of samples through a workflow. If actions that are marked with "Chain" are carried out on sample, then the Chain of Custody for the sample is updated as the sample moves through the workflow.

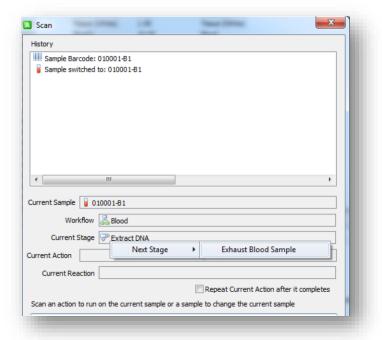
1. From any window that displays samples (Individuals window, Pedigrees window, Samples window, and so on), right-click on the sample (CTRL-click to select multiple samples), and on the context menu that opens, click Next Stage, and then select the appropriate workflow stage.

Figure 6-12: Moving a sample through a workflow from the Samples window



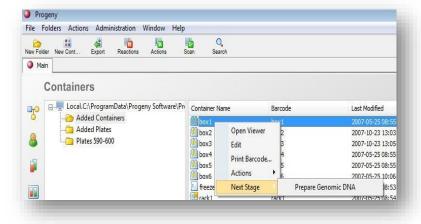
2. Open the Scan window, scan or enter the sample or container barcode, and then, right click in the Current Stage field, and on the Next Stage dropdown list that opens, select the appropriate stage for the samples.

Figure 6-13: Moving a sample through a workflow from the Scan window



- 3. On the Containers window, right-click on a container, and on the context menu that opens, click Next Stage, and then select the appropriate workflow stage.
  - a. Any action that is to be carried out on a container must be identified as a batch action.

Figure 6-14: Moving a sample batch through a workflow from the Containers window



## Appendix A: System Fields

System fields are read-only fields that Progeny automatically stores for a record. Each record type (individual, pedigree, sample, and marker) has its own unique system fields.

This appendix covers the following topics:

- <u>Individual System Fields</u>
- <u>Pedigree System Fields</u>
- <u>Sample System Fields</u>
- Marker System Fields

# Individual System Fields

Field Name	Description	Value Stored
Adopted	Defines an individual as adopted	0=Not Adopted; 1=Adopted In; 2=Adopted Out
Deceased Status	Defines an individual as deceased	1=True; 0=False
Degree of Relation <sup>1</sup>	Defines the degree of relation to the proband	0-infinity
Degree of Relation2 <sup>2</sup>	Defines the degree of relation to the proband	0-16
Father ID	Defines the UPN for the father of an individual	0-infinity
Folder	Lists the name of a folder that contains the individual	Name of folder
Folder Path	Lists the complete folder path to the individual including any parent folders	Full folder path
G: P	Generation number: Person number	1:1-infinity
Gender	Defines individual as male or female	M=male; F=Female
Gender Unknown	Defines individual gender as unknown	0=False;1=True
Genotyped	Defines if an individual is genotyped	0=No;1=Yes
Global ID	Unique number assigned to every individual in a database	1-infinity
Individual Created By	Progeny user who created the individual	User Name
Individual Created Date	Time/Date when the individual was created	Timestamp
Individual Last Modified	Time/Date when the individual was last modified	Timestamp
Individual Modified By	Progeny user who last modified the individual	User Name
Individual Name	Lists the name of the individual	Name of individual
Marked By	Defines individual marked by a sign	0=false, 1=+, 2=-, 3=*
Maternal-Paternal	Identified which side of the family an individual is part of	Mat, Pat, Both, None
Mother ID	Defines the UPN for the mother of an individual	0-infinity
No Issue	Defines an individual marked by No Issue	0=False;1=True
Pedigree Name	Lists the name of the pedigree that contains the individual	Name of pedigree
Primary Sample	Identifies the sample associated with an individual marked as "Primary"	Sample Name
Proband Status	Defines an individual as a proband	0=False;1=True

SAB	Defines an individual as still-born	0=False;1=True
Sample Count	Counts the number of samples for an individual	0-infinity
Twin Relationship	Lists the type of twin relationship for the individual	Monozygotic, Dizygotic, Unknown
Twin Status	Value for identifying twins	0=Not Twin;1=1st set, 3=2nd set; 5=3rd set, and so on
UPN	Lists the unique number of the individual in a pedigree	0-infinity

- 1. Defines the degree of relation of the specific individual to the proband. For example, a value of 1 is a first degree relative, the value of 2 is a second degree relative, and so on.
- 2. Defines a more specific relationship to the proband based on the following code system:

Code	Description
1	Proband
2	Brother or Sister
3	Son or Daughter
4	Parent
5	Paternal Grandparent
6	Paternal Aunt or Uncle
7	Maternal Grandparent
8	Maternal Aunt or Uncle
13	Nephew or Niece
14	Husband or Wife
15	Brother or Sister-in-Law
16	Identical Twin of Proband

# Pedigree System Fields

Field Name	Description	Value Stored
Audit Status	Indicates whether auditing is turned on for the pedigree	On or Off
FHQ Completed Date	Time/Date when FHQ is marked "Complete"	Timestamp
FHQ Last Invite Created By	Progeny user who sent most recent FHQ invite to pedigree	User Name
FHQ Last Invite Sent Date	Time/Date when most recent FHQ invite was sent	Timestamp
FHQ Num Invites Sent	Count of total FHQ invites sent to pedigree	0-infinity
FHQ Status	Indicates status of active FHQ	Invite Sent, In Progress, Complete
Folder	Lists the name of the folder that contains the pedigree	Name of folder
Folder Path	Lists the complete folder path to the pedigree including any parent folders	Full folder path
Last Modified	Lists the date when the pedigree was last modified.	Date of last modification
Modified By	Lists the name of the user who last modified the pedigree	Name of user
Pedigree Created By	Progeny user who created the pedigree	User Name
Pedigree Created Date	Time/Date when the pedigree was created	Timestamp
Pedigree Name	Lists the name of the pedigree	Name of pedigree

# Sample System Fields

Field Name	Description	Value Stored
Adopted	Defines an individual as adopted	0=Not Adopted; 1=Adopted In; 2=Adopted Out
Container Barcode	Barcode value representing Container Name	Box5
Container Name	Lists the name of the container for sample inventory	Box5
Container Path	Full path of specific container within container inventory	-20 Freezer\Shelf1\Box1
Container Type	Defines the type of container used	Building, Room, Freezer, Shelf, Rack, Box, Tank, Plate, Custom 1-6
Deceased Status	Defines an individual as deceased	1=True; 0=False
Degree of Relation <sup>1</sup>	Defines the degree of relation to the proband	0-infinity
Degree of Relation2 <sup>2</sup>	Defines the degree of relation to the proband	0-14
Father ID	Defines the UPN for the father of an individual	0-infinity
G: P	Generation number: Person number	1:1-infinity
Gender	Defines individual as male or female	M=male; F=Female
Gender Unknown	Defines individual gender as unknown	0=False;1=True
Global ID	Unique number assigned to every individual in a database	1-infinity
Individual Name	Lists the name of the individual	Name of individual
Marked By	Defines individual marked by a sign	0=false, 1=+, 2=-, 3=*
Maternal-Paternal	Identified which side of the family an individual is part of	Mat, Pat, Both, None
Mother ID	Defines the UPN for the mother of an individual	0-infinity
No Issue	Defines an individual marked by No Issue	0=False;1=True
Order Number	Unique identifier for orders (requires optional Orders Module)	0-infinity
Parent Sample Name	Lists the name of the parent sample for an aliquot	Name of parent sample
Pedigree Name	Lists the name of the pedigree that contains the individual	Name of pedigree
Position	Lists the location of the sample within a container	0-infinity
Proband Status	Defines an individual as a proband	0=False;1=True
SAB	Defines an individual as still-born	0=False;1=True
Sample Audit Status	Indicates whether auditing is turned on or off for the sample	On or Off

Sample Created By	Progeny user who created the sample	User Name
Sample Created Date	Time/Date when the sample was created	Timestamp
Sample Folder	Lists the name of the folder that contains the sample	Name of sample folder
Sample Last Modified	Time/Date when the sample was last modified	Timestamp
Sample Modified By	Progeny user who last modified the sample	User Name
Sample Name	Lists the name of the sample	Name of sample
Twin Relationship	Lists the type of twin relationship for the individual	Monozygotic, Dizygotic, Unknown
Twin Status	Value for identifying twins	0=Not Twin;1=1st set, 3=2nd set; 5=3rd set, and so on
UPN	Lists the unique number of the individual in a pedigree	0-infinity
Volume	Lists the current sample volume	0-infinity
Workflow	Lists the name of the workflow with which the sample is associated	Name of workflow
Workflow stage	Lists the name of the workflow stage that was last completed for the sample	Name of workflow stage

- 1. Defines the degree of relation of the specific individual to the proband. For example, a value of 1 is a first degree relative, the value of 2 is a second degree relative, and so on.
- 2. Defines a more specific relationship to the proband based on the following code system:

Code	Description
1	Proband
2	Brother or Sister
3	Son or Daughter
4	Parent
5	Paternal Grandparent
6	Paternal Aunt or Uncle
7	Maternal Grandparent
8	Maternal Aunt or Uncle
13	Nephew or Niece
14	Husband or Wife
15	Brother or Sister-in-Law
16	Identical Twin of Proband

# Marker System Fields

Field Name	Description	Value Stored
Base Pairs	The value entered for base pairs	0-infinity
Centimorgans	Value entered for Centimorgans	0-infinity
Chromosome Name	Name of the chromosome where the marker is located	0-infinity
Marker Name	List name of the microsatellite or SNP	Name of marker
Marker Type	Defines the type of marker	Marker or SNP
AA Count	Count of AA calls per SNP	0-infinity
AB Count	Count of AB calls per SNP	0-infinity
BB Count	Count of BB calls per SNP	0-infinity
NoCall Count	Count of NoCalls per SNP	0-infinity
Call Rate	Comparison of AA + AB + BB calls versus NoCalls per SN: P	0-infinity
Degree of Heterozygosity	The proportion of heterozygotes per SNP	0-1
Hardy-Weinberg P-Value	P-value results for Hardy Weinberg tests per SNP	0-1
Minor Allele Frequency	The ratio of the allele that is less common to the total number of alleles (2) per SNP	05
Minor Allele	Stores the minor allele	A or B
Allele A Value	Stores the expected value for Allele A	ACG or T
Allele B Value	Stores the expected value for Allele B	ACG or T

## Appendix B: Importing Clinical Data

You use the Import module to import external pedigrees, individuals, and samples that do not already exist in a Progeny database. You can also use the Import module to import data that updates individuals and samples that already exist in the database. The data can be contained in any non-complex field (not a computed field, not a summary field, and so on) other than a table field. If you import pedigrees that do not already exist in the database, Progeny automatically draws the pedigree and imports all the associated data fields as well.



If you need to import table data for pedigrees, individuals, or samples, you must use the spreadsheet import function. See <u>To import data into a spreadsheet</u>

This appendix covers the following topics:

- Overview of the Import Module
- Running the Import Module

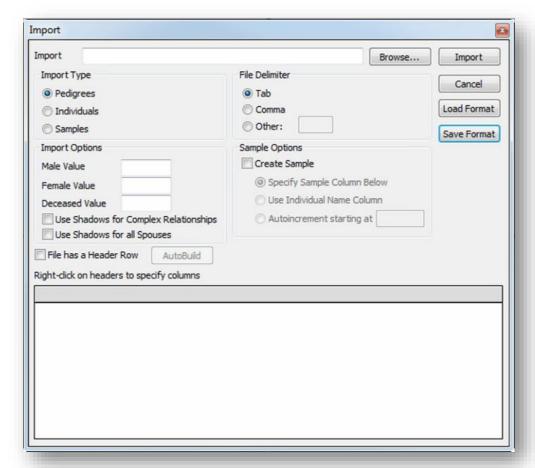
# Overview of the Import Module

You use the Import module to import three types of external data that do not already exist in a Progeny database – pedigrees, individuals not contained in pedigrees, and samples. You can also use the Import module to import data that updates individuals and samples that already exist in the database. The data can be contained in any non-complex field (not a computed field, not a summary field, and so on) other than a table field. If you import pedigrees that do not already exist in the database, Progeny automatically draws the pedigree and imports all the associated data fields as well. If you import pedigrees and individuals, you are importing individual-level data fields. If you import samples, you are importing sample level data fields.



If you need to import table data for pedigrees, individuals, or samples, you must use the spreadsheet import function. See <u>To import data into a spreadsheet</u>

Figure B-1: Import module dialog box (for pedigrees)



All pedigree data and all individual data must be contained in separate text files that have a specific structure. The sample text file can contain be imported as a standalone file, or the sample data can be attached to a pedigree or an individual. Any type of import file can contain fields that exist in the Progeny database as well as fields that do not exist in the Progeny database. If the fields do not exist in the Progeny database, then the fields must be of the same type – that is, they must all be text fields, must all be number fields, and so on. After you import the file, if needed, you can change the field type for these undefined fields. The import options that are available on the Import dialog box depend on whether you are importing pedigree data, individual data, or sample data.

### Import file structure for pedigrees

Five columns of the pedigree text file must contain data that defines the pedigree structure—Pedigree Name, Individual Name, Gender, Mother ID, and Father ID. The remaining columns can contain relationship information (spouse, twin status, sibling, and so on) and then individual information (first name, last name, date of birth, and so on).

### Pedigree Import Required Columns

- Pedigree Name Unique alphanumeric ID for the pedigree name.
- Individual Name Unique alphanumeric ID for the individual name. (Must be unique per pedigree)
- Father ID Unique alphanumeric ID for the father name.
- Mother ID Unique alphanumeric ID for the mother name.
- Gender Distinction between male and female. (If the field is blank or uses a value that is or different from the Male/Female values, gender is marked as Unknown.)

### Pedigree Import Optional Columns

- Child Unique alphanumeric ID for the child name.
- Sibling Unique alphanumeric ID for the sibling name.
- Spouse 1 Unique alphanumeric ID for the first spouse name.
- Spouse 2 Unique alphanumeric ID for the second spouse name.
- Twin Status Number used to identify a set of twins. For instance, the first set of twins within a pedigree would all have the number 1. The second set of twins would have the number 2, and so on.
- Twin Relationship Twin designation: monozygotic or dizygotic
- Proband Status If your proband is not the first person listed for each pedigree, designate the correct proband in this column with the value of 1.
- Deceased Value specified in Import Options on the Import-Module dialog box for pedigrees.
- Sample Name Alphanumeric ID for sample name if "Create Sample/Specify Sample Column Below" is selected in Sample Options on the Import-Module dialog box for pedigrees.

## Import file structure for individuals

Two columns of an individual text file must contain the individual name (or ID) and gender. The remaining columns can contain any other information for the individual such as first name, last name, date of birth, and so on.

### *Individual Import Required Columns*

- Individual Name Alphanumeric ID for everyone. (Must be unique per individual.)
- Gender Distinction between male and female. (If the field is blank or uses a value that is or different from the Male/Female values, gender is marked as Unknown)

### Individual Import Optional Columns

- Deceased Value specified in Import Options on the Import-Module dialog box for individuals.
- Sample Name Alphanumeric ID for sample name if "Create Sample/Specify Sample Column Below" is selected in Sample Options on the Import-Module dialog box for individuals.

### Import file structure for samples

One column of a sample text file must contain the sample name (or ID). The remaining columns can contain other relevant information about the sample such as container name, barcode, and so on.

### Sample Import Required Column

• Sample Name - Alphanumeric ID for the sample name. (Must be unique per sample.)

#### Sample Import Optional Columns

- Container Name Allows you to place a sample into a container that already exists in the database
- Barcode Unique label ID assigned to a sample for tracking its location in a container.
- Volume Numeric value for the quantity of available sample.
- Parent Sample Name Alphanumeric ID of the original sample from which the aliquot was derived.
- Workflow Name Allows you to place a sample into a workflow that already exists in the database.
- Position Exact location within the container.

# Running the Import Module

You can run the Import module from the Pedigrees window, the Individuals window, or the Samples window. The type of data that is selected for importing and the folders in which the data is imported all depend on the window from which you opened the Import module.

### To run the import module

- 1. Open the Pedigrees, Individuals, or Samples window and select a folder on the window.
- 2. Click the Import button Import on the Pedigrees, Individuals, or Samples window toolbar.
  - a. If you clicked the Import button from...
    - i. ...the Pedigrees window, then Pedigrees is selected for the Import Type.
    - ii. ...the Individuals window, then Individual is selected for the Import Type.
    - iii. ...the Samples window, then Samples is selected as the Import Type.

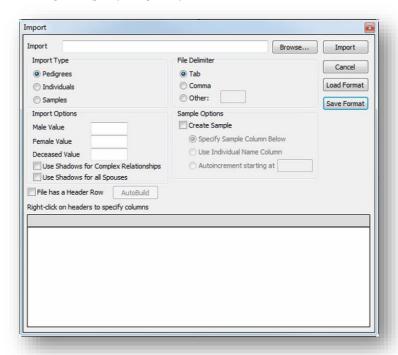


You can, however, import any of the three types of data from an access point.

- b. If you opened the Import module from the Pedigrees window, then the pedigree data is imported into the pedigree folder that you selected on the Pedigrees window. If you opened the Import module from either the Individuals or Samples window and selected Pedigrees as the Import Type, then the pedigree data is imported into a new folder (Imported Pedigrees) that is stored at the root level for the Individuals or Samples window.
- c. If you opened the Import module from the Individuals window, then the individual data is imported into the individual folder that you selected on the Individuals window. If you opened the Import module from either the Pedigrees or Samples window and selected Individual as the Import Type, then the individual data is imported into a new folder (Imported Individuals) that is stored at the root level for the Pedigrees or Samples window.
- d. If you opened the Import module from the Samples window, then the sample data is imported into the sample folder that you selected on the Samples window. If you opened the Import module from either the Pedigrees or Individuals window and selected Sample as the Import Type, then the sample data is imported into a new folder (Imported Samples) that is stored at the root level for the Pedigrees or Individuals window.
- 3. Continue to one of the three data type imports detailed below...

## To import pedigree data

Figure B-2: Import module dialog box (for pedigrees)



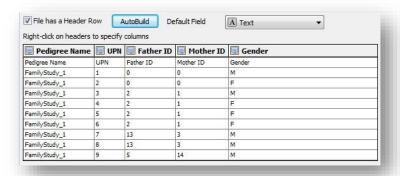
- 1. Click Browse to browse to and select the data file that is being imported. The imported file is displayed in the Import Preview pane (bottom pane) of the dialog box.
- 2. Select the delimiter that is used for the data in the import file. If you select Other, you must specify the character that is used for the delimiter.
- 3. Specify the Import Options for the file:
  - a. Male Value Specify the value used that is to be used for males in the Gender column in your file (for example, 1, M, Male).
  - b. Female Value Specify the value that is to be used for females in the Gender column in your file (for example, 2, F, Female).
  - c. Deceased Value Specify the value that is to indicate deceased in the Deceased column in your file (for example, 1, D).
  - d. Use Shadow for Complex Relationships Shadow is a feature that allows you to a show a complex marital relationship that otherwise would be shown as a single line that would cross the entire pedigree.
  - e. Use Shadows for all Spouses Creates shadows for all spouses that cannot be easily connected.

- 4. If sample data is to be imported with the pedigree data, select Create Sample, and then specify the Sample Options for the file.
  - a. Specify Sample Column Below Select this option if you are specifying a column that contains your Sample Name in the import preview.
  - b. Use Individual Name column Select this option if the newly created samples will use the name from the Individual Name column that is specified in the import preview.
  - c. Auto increment starting at Select this option to create a sample for everyone in the pedigree and auto increment the sample number starting at the number specified.
- 5. In the Import Preview pane (the bottom pane of the Import dialog box), do one of the following:
  - a. If your import file has a header row, then select File has Header Row, and then click AutoBuild. Also, if your import file contains fields that do not currently exist in the Progeny database, select the field type on the Default Field dropdown list. (The default value is Text.)
  - b. Progeny looks at the header row in your import file and determines what the appropriate header should be for each column in the data file. (See <u>Figure B-3</u>) If your import file contains fields that do not exist in the Progeny database, Progeny creates these fields in the Progeny database, assigns the headers that are in the import file as the column headers, and imports the data that is in each column into the newly created field.
  - c. If your import file does not have a header row, then you must right-click in the header for each column and manually assign the appropriate column heading. (See <u>Figure B-4</u>) If yours contains fields that do not exist in the Progeny database, then continue to Step 6; otherwise, continue to Step 7.



If you do not agree with an automatic heading that Progeny has assigned to a column, you can also use the manual option to assign a different heading.

Figure B-3: Import file with a header row





After you select File has Header Row, and then click AutoBuild, Progeny looks at the header row in your import file and determines what the appropriate header should be for each column in the data file.

Figure B-4: Manually assigning a column heading



- 6. For each field that is not contained in the Progeny, right-click in the field, and then do one of the following:
  - a. To have the data imported into a new database field, select Create New Field.
    - i. The Add a new Field dialog box opens. Enter a name for the new field in this dialog box, and optionally, select a different field type. The field will be created and the data for the individual will be imported into this field during the import process.

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Figure B-5: Add a New Field dialog box





Remember, when you import pedigree data, you are importing individual-level data fields. As a result, after the import process is complete, this field can be found in an individual data fields folder named Import Fields in the Fields window.

- b. To have the data imported into an existing database field, select Choose Existing Field.
  - i. The Select Field dialog box opens, from which you can select an existing individual database field into which to import the data.

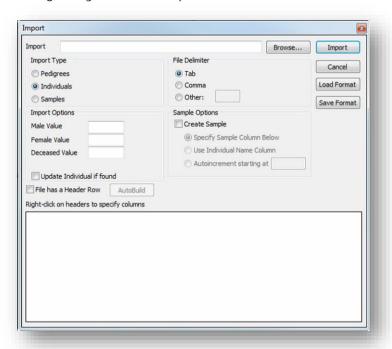
Figure B-6: Select Field dialog box



- 7. Optionally, after you label a column, you can do one or both of the following:
  - a. To remove a column from the import, right-click on the column, and on the context menu that opens, select Ignore This Column.
  - b. To update a column a column after it has been labeled, right-click on the column, and on the context menu that opens, select a different column label.
- 8. Click Import. A Progeny Import Module dialog box opens, indicating the status of the import.
- 9. When the import is complete, click Close.

## To import individual data

Figure B-7: Import module dialog box (for individuals)



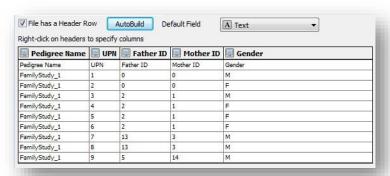
- 1. Click Browse to browse to and select the data file that is being imported. The imported file is displayed in the Import Preview pane (bottom pane) of the dialog box.
- 2. Select the delimiter that is used for the data in the import file. If you select Other, you must specify the character that is used for the delimiter.
- 3. Specify the Import Options for the file:
  - a. Male Value Specify the value used that is to be used for males in the Gender column in your file (for example, 1, M, Male).
  - b. Female Value Specify the value that is to be used for females in the Gender column in your file (for example, 2, F, Female).
  - c. Deceased Value Specify the value that is to indicate deceased in the Deceased column in your file (for example, 1, D).
  - d. Update Individual if Found—Select this option to update the field values for an individual if the individual is found to already exist in the Progeny database.

- 4. If sample data is to be imported with the individual data, select Create Sample, and then specify the Sample Options for the file.
  - a. Specify Sample Column Below Select this option if you are specifying a column that contains your Sample Name in the import preview.
  - b. Use Individual Name column Select this option if the newly created samples will use the name from the Individual Name column that is specified in the import preview.
  - c. Auto increment starting at Select this option to create a sample for everyone in the pedigree and auto increment the sample number starting at the number specified.
- 5. In the Import Preview pane (the bottom pane of the Import dialog box), do one of the following:
  - a. If your import file has a header row, then select File has Header Row, and then click AutoBuild. Also, if your import file contains fields that do not currently exist in the Progeny database, select the field type on the Default Field dropdown list. (The default value is Text.)
    - i. Progeny looks at the header row in your import file and determines what the appropriate header should be for each column in the data file. (See <u>Figure B-8</u>) If your import file contains fields that do not exist in the Progeny database, Progeny creates these fields in the Progeny database, assigns the headers that are in the import file as the column headers, and imports the data that is in each column into the newly created field. Continue to Step 7.
  - b. If your import file does not have a header row, then you must right-click in each column header and manually assign the appropriate column heading. (See <u>Figure B9</u>) If yours contains fields that do not exist in the Progeny database, then continue to Step 6; otherwise, continue to Step 7.



If you do not agree with the automatic heading that Progeny has assigned to a column, you can also use the manual option to assign a different heading.

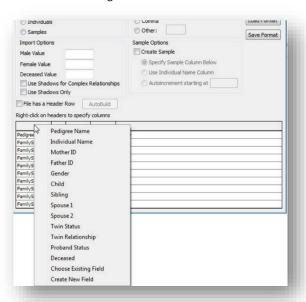
Figure B-8: Import file with a header row





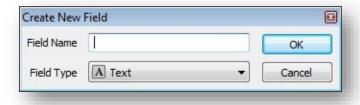
After you select File has Header Row, and then click AutoBuild, Progeny looks at the header row in your import file and determines what the appropriate header should be for each column in the data file.

Figure B-9: Manually assigning a column heading



- 6. For each field that is not contained in the Progeny database, right-click in the field, and then do one of the following:
  - a. To have the data imported into a new database field, select Create New Field.
    - i. The Add a new Field dialog box opens. Enter a name for the new field in this dialog box, and optionally, select a different field type. The field will be created and the data for the individual will be imported into this field during the import process.

Figure B-10: Add a New Field dialog box





After the import process is complete, this field can be found in an Individual Data Fields folder named Import Fields in the Fields window.

- b. To have the data imported into an existing database field, select Choose Existing Field.
  - i. The Select Field dialog box opens, from which you can select an existing individual database field into which to import the data.

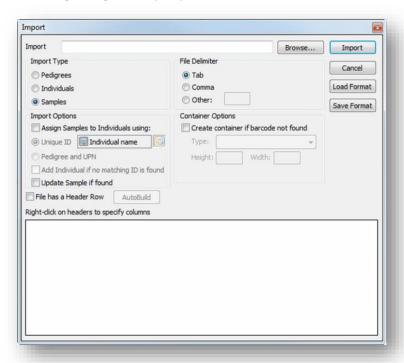
Figure B-11: Select Field dialog box



- 7. Optionally, after you label a column, you can do one or both of the following:
  - a. To remove a column from the import, right-click on the column, and on the context menu that opens, select Ignore This Column.
  - b. To update a column a column after it has been labeled, right-click on the column, and on the context menu that opens, select a different column label.
- 8. Click Import. A Progeny Import Module dialog box opens, indicating the status of the import.
- 9. When the import is complete, click Close.

## To import sample data

Figure B-12: Import module dialog box (for samples)



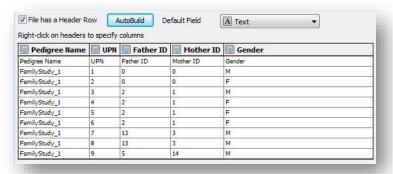
- 1. Click Browse to browse to and select the data file that is being imported. The imported file is displayed in the Import Preview pane (bottom pane) of the dialog box.
- 2. Select the delimiter that is used for the data in the import file. If you select Other, you must specify the character that is used for the delimiter.
- 3. Specify the Import Options for the file:
  - a. Unique ID Specify the Unique ID field used to connect your new samples with an existing individual in the database.
  - b. Pedigree and UP Connect your new samples with a Pedigree and UPN combination that already exists in the database.
  - c. Add Individual if no matching ID is found Select this option to add a new individual if one of the previous matching criteria is not met.
  - d. Update Sample if Found—Select this option to update the field values for a sample if the sample is found to already exist in the Progeny database.

- 4. In the Import Preview pane (the bottom pane of the Import dialog box), do one of the following:
  - a. If your import file does have a header row, then select File has Header Row, and then click AutoBuild. In addition, if your import file contains fields that do not currently exist in the Progeny database, select the field type on the Default Field dropdown list. (The default value is Text.)
    - i. Progeny looks at the header row in your import file and determines what the appropriate header should be for each column in the data file. (See <u>Figure B-13</u>) Also, if your import file contains fields that do not exist in the Progeny database, Progeny creates these fields in the Progeny database, assigns the headers that are in the import file as the column headers, and imports the data that is in each column into the newly created field. Continue to Step 6.
  - b. If your import file does not have a header row, then you must right-click in each column header and manually assign the appropriate column heading. (See <u>Figure B14</u>) If yours contains fields that do not exist in the Progeny database, then continue to Step 5; otherwise, continue to Step 6.



If you do not agree with the automatic heading that Progeny has assigned to a column, you can also use the manual option to assign a different heading.

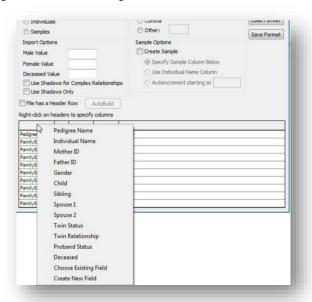
Figure B-13: Import file with a header row





After you select File has Header Row, and then click AutoBuild, Progeny looks at the header row in your import file and determines what the appropriate header should be for each column in the data file.

Figure B-14: Manually assigning a column heading



- 5. For each field that is not contained in the Progeny, right-click in the field, and then do one of the following:
  - a. To have the data imported into a new database field, select Create New Field.
    - i. The Add a new Field dialog box opens. Enter a name for the new field in this dialog box, and optionally, select a different field type. The field will be created and the data for the individual will be imported into this field during the import process.

Figure B-15: Add a New Field dialog box

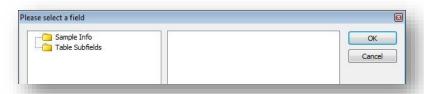




After the import process is complete, this field can be found in a Sample Data Fields folder named Import Fields in the Fields window.

- b. To have the data imported into an existing database field, select Choose Existing Field.
  - i. The Select Field dialog box opens, from which you can select an existing individual database field into which to import the data.

Figure B-16: Select Field dialog box



- 6. Optionally, after you label a column, you can do one or both of the following:
  - a. To remove a column from the import, right-click on the column, and on the context menu that opens, select Ignore This Column.
  - b. To update a column a column after it has been labeled, right-click on the column, and on the context menu that opens, select a different column label.
- 7. Click Import. A Progeny Import Module dialog box opens, indicating the status of the import.
- 8. When the import is complete, click Close.

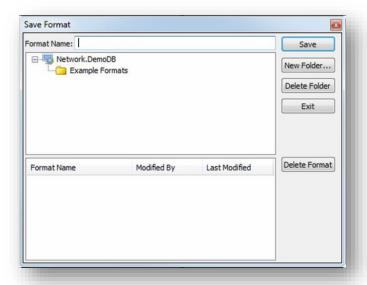
# Saving and Loading an Import Format

After you have set up an import format for pedigrees, individuals, or samples, you can save the format. You can then load this saved format and run it on an as-needed basis.

### To save an import format

- 1. Set up the import format for the pedigrees, individuals, or samples, but do not click Import. See:
  - a. To import pedigree data
  - b. To import individual data
  - c. To import sample data
- 2. Click Save Format on the Import dialog box.
- 3. The Save Format dialog box opens. It provides a default folder named Progeny in which to save the import format.

Figure B-17: Save Format dialog box



- 4. Enter a name for the import format, and then do one of the following:
  - a. Select the Progeny folder.
  - b. Click New Folder, and on the Format Folder dialog box that opens, do the following:
    - i. Enter the name of the new folder in which to save to format.
    - ii. Select the location for the new folder. The folder can be stored at the root level (the same level as the Progeny folder), or it can be a sub-folder of the Progeny folder, or of another folder. Click Save.
- 5. Click Save on the Save Format dialog box. You can then load this saved format and run it on an as-needed basis.

### To load an import format

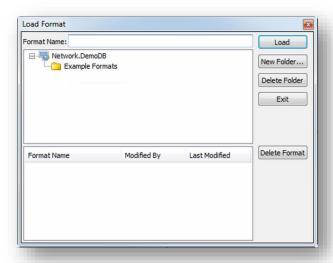
You can a retrieve a saved import format and load it to import a data file with the appropriate type of data. For example, if you retrieve a saved pedigree import, then you must select a text file that contains pedigree data for importing.

1. Open the Import module.



2. On the Import dialog box, click the Load Format button Load Format dialog box opens.

Figure B-18: Load Format dialog box



- 3. Open the folder that contains the import format that you are loading, select the format then click Load.
- 4. The Import dialog box is refreshed with the loaded format. You can now browse and select the import file, and then click Import to import the data according to the selected format.

# Appendix C: Progeny Workflows Client Interface

## What is the Workflow Interface and how is it different?

The Progeny Workflow Interface has been designed as a simplified version of the classic Progeny 9 user interface. This has been developed as a direct response to our customer base wanting an "easy mode" for Progeny that could both allow new users to quickly acclimate to the Progeny environment and restrict limited-access users to the specific areas of Progeny they need to accomplish their work. Consisting of seven different workflow-based "apps" this interface can be activated within the User Maintenance menu and each of the "apps" can be individually assigned to a user. This document will detail the use/functionality for each of the seven workflow apps and provide instructions for Progeny administrators on how to activate and configure the interface.



The first six workflow apps (New Pedigree, Review Pedigree, New Individual, Update Individual, Build Spreadsheet and Run Spreadsheet) can be used on both the desktop and web client platforms. However, the New Questionnaire workflow app is only available within the Progeny web client and requires the Family History Questionnaire module to be activated to properly function.

## Add New Pedigree Workflow

This app allows a user to create a new pedigree. Progeny 8 web client users will recognize the steps in this workflow as it uses many of the components from Progeny 8's Add Family button. The first step of the workflow asks the user to choose a folder – the administrator can define a default folder for this step, and as seen in the screenshot on the next page the Pedigrees folder is already selected. Users can choose a specific folder or accept the default folder selection then click the Next button.



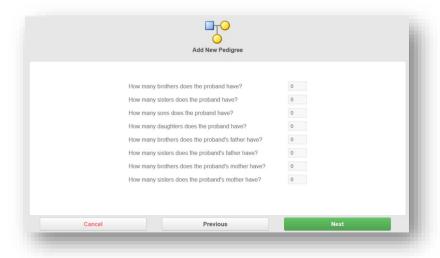
The next step prompts the user to enter to enter a name for the new pedigree, then click the Next button. This step validates the pedigree name against the rest of the database, so if the entered pedigree already exists the user will receive a warning and be forced to enter a new pedigree name.



Next the user will be prompted to choose the gender for the proband – however, the Unknown gender option is not available to users from the workflow interface.

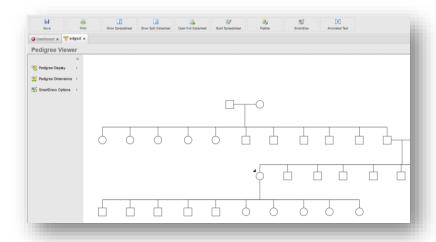


The last step of the workflow prompts the user to enter the number of first and second-degree relatives within the proband's family. Users can click on each field to enter a number <u>or</u> use the Tab key.



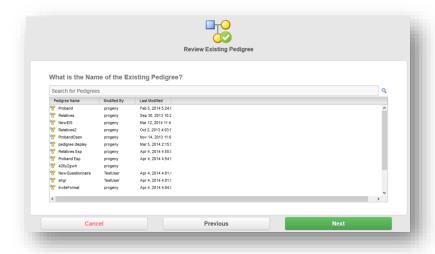
Once the number of family members has been entered, the user will click Next and Progeny will perform the following steps automatically:

- 1. Create the Pedigree
- 2. Add the Proband and assigns the chosen gender
- 3. Add relatives based on the numbers entered in the previous step
- 4. Save the pedigree then open the pedigree viewer



## Review Existing Pedigree Workflow

When this workflow app is activated, a search box will open providing the user an opportunity to either browse the pedigrees in a combined list or use the Search for Pedigrees bar at the top of the window to perform a full-text search across the pedigree names. Users can enter a full or partial pedigree name and Progeny will return a list of all pedigrees whose name contains the text string entered the search box. Once the user chooses a pedigree and clicks the Next button, Progeny will launch the pedigree viewer for the selected pedigree.



### Add New Individual Workflow

This workflow is almost identical to the Add New Pedigree workflow in its functionality. The first step for the user will be to choose the folder where the individual is to be created. The user can accept the configured default folder or choose their desired folder then click the Next button to continue.



Next, the user will enter the name of the new individual and click the Next button when finished.



The last step for the user is to choose the gender of the new individual – again, the Unknown gender option is not available within the workflow. Once a gender is selected the user will click the Next button and Progeny will create the individual open the Individual Datasheet.



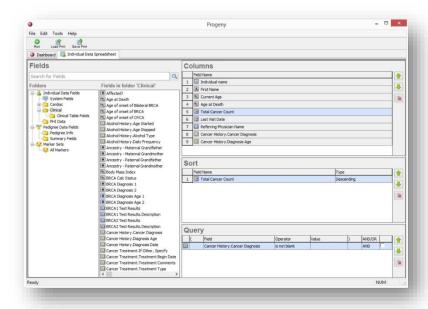
## Update Existing Individual Workflow

This workflow app is identical in function to the Review Existing Pedigree workflow. When activated, a search box will open providing the user an opportunity to either browse the individuals in a combined list or use the Search for Individuals bar at the top of the window to perform a full-text search across the individual names. Users can enter a full or partial individual name and Progeny will return a list of all individuals whose name contains the text string entered in the search box.



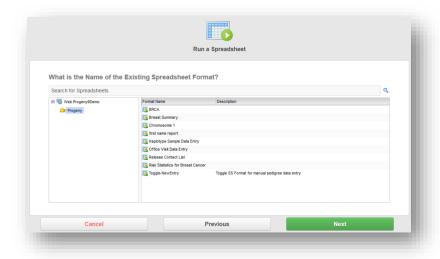
## **Build Spreadsheet Workflow**

This workflow opens the build menu for the Individual Data Spreadsheet. Please refer to the Progeny 9 User Guide for instructions on how to build an Individual Data Spreadsheet.



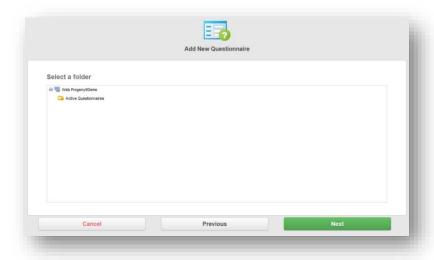
## Run a Spreadsheet Workflow

This workflow will open the Spreadsheet Formats window, allowing the user to either search for a spreadsheet format or browse the available saved spreadsheet formats. Once a format has been selected the user clicks the Next button then Progeny will immediately run the saved spreadsheet.



### Add New Questionnaire Workflow

This workflow is only accessible through the Progeny web client and requires the Family History Questionnaire module to function properly. The Add New Questionnaire Workflow is especially useful for limited-access users such as schedulers and interns as an administrator can set up a user account that has this (and only this) workflow. After selecting the Add New Questionnaire workflow button, the user will select a folder then click the Next button. **Only folders that are activated for the Family History Questionnaire will display in the folder list.** 



In the second step, the user will enter a name for the pedigree then click the Next button.



Next, the user will choose the gender of the proband. Once again, the Unknown gender option is not available within the workflow.



Step 4 has the user entering the patient's email address. This is required to send the patient an email invitation to take the Family History Questionnaire.



In the last step, the user is presented with a page of optional additional fields to fill in for the patient. This page can be customized to display the additional fields an administrator deems relevant. To create the layout for this page, the administrator will build a single-tab Individual Datasheet then save this as a datasheet format. Within the Family History Questionnaire configuration, this saved datasheet format will be entered in the Invite Datasheet Format option within the Configurable Options list.



When the user clicks the Next button on this page, the Progeny application will perform the following steps automatically:

- 1. Create the pedigree
- 2. Add the proband
- 3. Insert data into additional fields from the current workflow page
- 4. Generate the email invitation and send it to the patient

When the process completes, and the email successfully sends, the user will be given a confirmation message. Once this message is closed the workflow will complete and close.

# Configuring User Accounts for Workflow User Interface

To create a new user account with the workflow interface or to modify an existing user account to use the workflow interface, log into your Progeny database as the 'progeny' Superuser then click Administration  $\rightarrow$  User Maintenance. Create the new (or select the existing) user account then in the bottom half of the menu select the Client Interface tab.

The two main options available are Use Full Progeny Client (current Progeny Interface) and Use Dashboard Client (new Workflows Interface). Click the radio button for Use Dashboard Client then select the checkboxes from the available list to assign workflow apps to the user account. For the New Pedigree and New Individual apps, a default folder can be optionally assigned.

