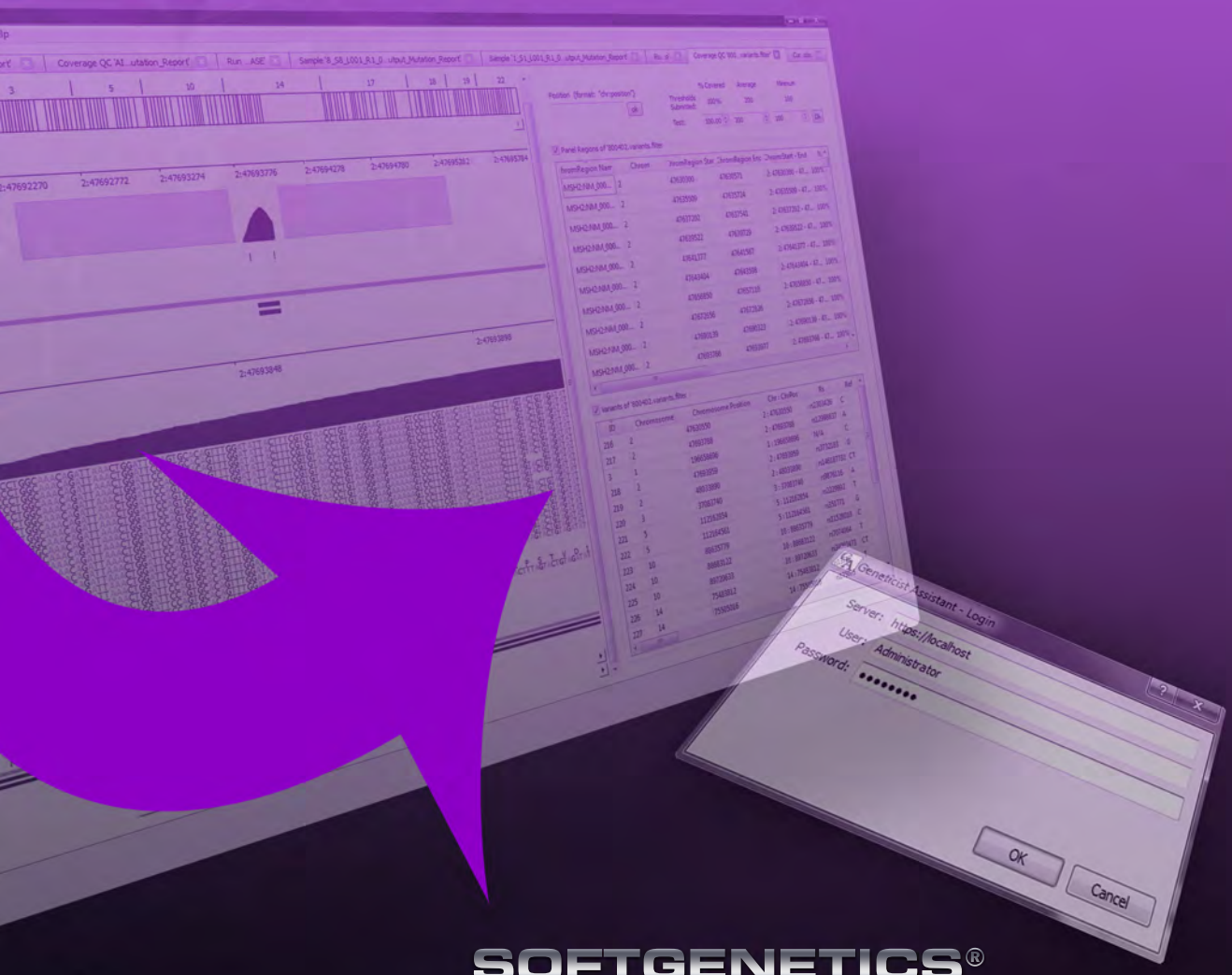
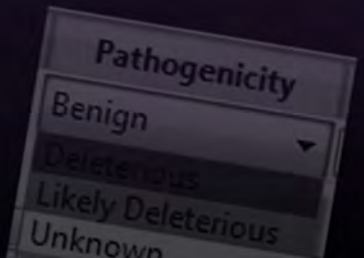
GeneticistAssistant™

NGS Interpretative Workbench

User Manual





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Release Information

Document Version Number	*GeneticistAsst-1.1.6-UG002*
Software Version	*1.1.6*
Document Status	*Final*

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Table of Contents

Preface

Welcome to the *Geneticist Assistant User's Manual*. The purpose of the *Geneticist Assistant User's Manual* is to answer your questions and guide you through the procedures necessary to use the Geneticist Assistant application efficiently and effectively.

Using the manual

You will find the *Geneticist Assistant User's Manual* easy to use. You can simply look up the topic that you need in the table of contents or the index. Later, 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 *Geneticist Assistant 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 *Geneticist Assistant 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, <Gene 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.

Although numbering begins on the cover page, this number is not visible on the cover page or front matter pages. Page numbers are visible beginning with the first page of the table of contents.

- 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 *Geneticist Assistant User's Manual* assumes that:

- You are familiar with windows-based applications and basic Windows functions and navigational elements.
- References to any third party standards or third party software functions were current as of the release of this version of Geneticist Assistant, and might have already changed.

Organization of the manual

In addition to this Preface, the *Geneticist Assistant User's Manual* contains the following chapters and appendices:

- Chapter 1, "Getting Started with Geneticist Assistant," on page 11 details the installation requirements and installation procedure for Geneticist Assistant. It also explains how to open and log into Geneticist Assistant and it provides an overview of the major navigational elements on the main window for the application, the main menu, the toolbar, the tabs display area, and the status bar.
- Chapter 2, "Geneticist Assistant Setup and Maintenance," on page 39 details the objects that are available in Geneticist Assistant and how you can add, edit, or delete values for these objects. It also details the various settings (reference directory, pathogenicity colors coverages, variant and tab preferences, Alamut, HGMD, user password, OMIM, and

license server) that you can specify for your Geneticist Assistant installation. Finally, it explains how to back up the Geneticist Assistant database.

- Chapter 3, “Geneticist Assistant User Management,” on page 63 details how to manage users and groups in Geneticist Assistant, including adding new users and groups, deleting users and groups, and managing the permissions for users and groups.
- Chapter 4, “Managing Panels,” on page 77 details the management of your Geneticist Assistant panels, which includes, adding, modifying and deleting panels, and reviewing panel statistics.
- Chapter 5, “Managing Patients and Runs,” on page 93 details the management of patients. It also details how to add and carry out runs in Geneticist Assistantt.
- Chapter 6, “Geneticist Assistant Data Review,” on page 129 details all the options that are available to you for reviewing the data that is generated by a run.
- Chapter 7, “Geneticist Assistant Reports,” on page 203 details the generation of the six standard reports that are available in Geneticist Assistant. It also details how to generate a custom report, which includes only the data that you select.
- Chapter 8, “Geneticist Assistant Tools,” on page 213 details the various tools that are available in Geneticist Assistant to assist you with setting up, running, and analyzing sample data.
- The Glossary on page 225 provides a detailed definition of specialized terms for Geneticist Assistant.

Preface

Chapter 1
Getting Started with Geneticist Assistant

Geneticist Assistant is a web-based tool for the control, visualization, interpretation, and creation of a historical knowledge base of next generation sequencing data. This data is used to identify potentially pathogenic variants that are associated with specific conditions such as hereditary colon cancer. This chapter details the installation recommendations for Geneticist Assistant and how to install Geneticist Assistant. It also explains how to launch Geneticist Assistant and provides an overview of the major navigational elements for the application, including the menu bar, the toolbar, and the main window tabs.

This chapter covers the following topics:

Chapter 1
Getting Started with Geneticist Assistant

Geneticist Assistant System Recommendations

You must have Administrator rights for the computer on which you are installing Geneticist Assistant.

Table 1-1: Geneticist Assistant Recommended Hardware Specifications

Client and/or Server	
Operating System	• Windows 7, 64 bit (Recommended) • Windows Vista (64 bit) • Windows Server 2008 (64 bit) • Windows Server 2012 (64 bit)
Processors	Two cores (64 bit)
RAM	2 GB
Hard Drive	100 GB (Minimum)
Note: Virtual Machines are supported.	

Table 1-2: Geneticist Assistant Data Specifications

Database	Client
• 1 GB disk space	• 50 GB space for reference and annotation data
• Starts at 15 MB and grows slowly	• BAM files - Ca. the same size as raw FASTQ data
• Backup with mysqldump	
• Low IOPS	

Table 1-3: Geneticist Assistant Software Details

Server	Client
• Apache 2.2 (Latest)	• Qt 4.84
• mod_wsgi (Latest)	• Bamtools 2.1.1
• OpenSSL (Latest)	• Zlib 1.2.8
• MySQL 5.6 (Latest)	• Qjson 2.1
• Python 2.7 (Latest) • Django 1.6 (Latest) • South (Latest) • Setuptools (Latest) • MySQLdb (Latest)	• OpenSSL 0.9.7

Installing the SoftGenetics License Server

Before you install Geneticist Assistant, if you have a *purchased* version of Geneticist Assistant, then you must install the SoftGenetics license server. The *license server* is a central location either locally or on a network where software application licenses are stored and can be accessed by users of the application. After you install and open Geneticist Assistant for the first time, you will be prompted to configure Geneticist Assistant to connect to the SoftGenetics license server so that you can use Geneticist Assistant. The executable to install the SoftGenetics license server is named SrvSetup_<>_exe and it is available from SoftGenetics's ftp site. Contact SoftGenetics for assistance in obtaining this file.

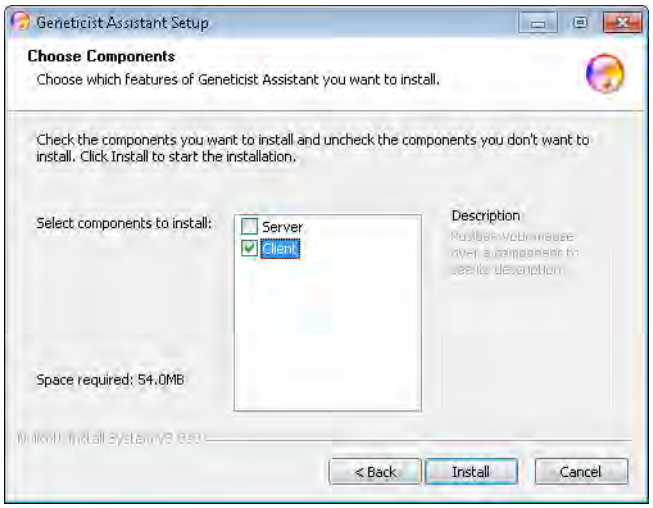
If you are running a trial version of Geneticist Assistant, then you do not need to set up the SoftGenetics license server. You can simply indicate that you are running a trial version after you log into Geneticist Assistant.

Installing Geneticist Assistant

SoftGenetics provides a single standard executable file—Geneticist AssistantSetup< >.exe—for the installation of both the Geneticist Assistant client and the database server. You must have Administrator rights on the computer(s) on which you are installing Geneticist Assistant. You can install the client and the server on the same computer or on different computers. When the installer first opens, Server is *not* selected and Client is selected.

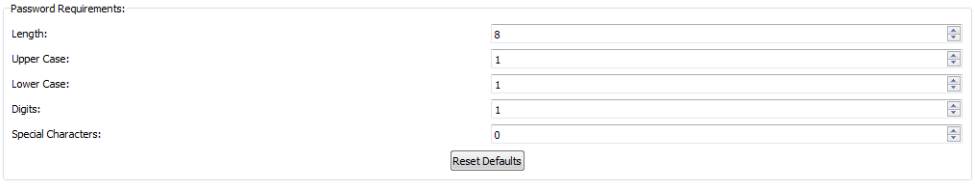
- If you are installing the client and database server on the *same* computer, then you must select Server before you click Install.
- If you are installing the client and database server on *different* computers, then:
  - When you run the installer on the computer on which you are installing the Geneticist Assistant client, you must leave the Server option cleared and the Client option selected.
  - When you run the installer on the computer on which you are installing the database server, you must select the Server option and clear the Client option.

Figure 1-1: Geneticist Assistant Installation wizard, Client/Server options



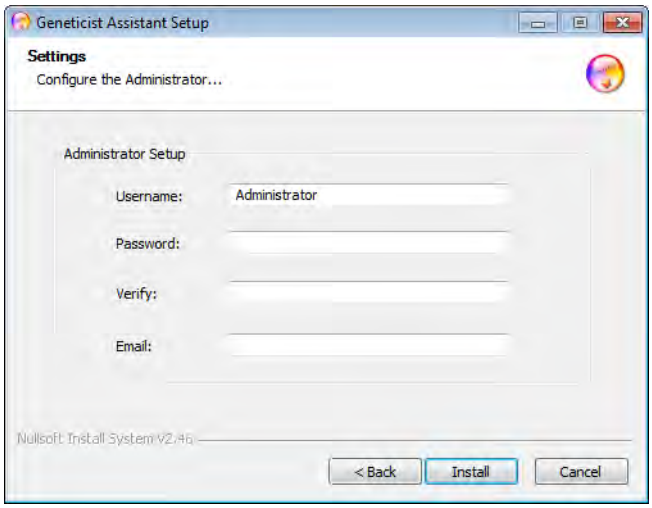
When you install Geneticist Assistant, you are prompted to create the Administrator role for the installation. By default, the Username is set to Administrator, but you can always enter a different value. The password cannot contain any spaces. If you intend to implement additional password requirements, then the password must also adhere to these planned requirements. Figure 1-2 on page 16 shows the requirements that you can specify for user passwords after Geneticist Assistant is installed. (See “To specify the Geneticist Assistant password requirements” on page 57.)

Figure 1-2: User password requirement options



The email address is the address that is linked to the Administrator account.

Figure 1-3: Administrator Setup dialog box



The Geneticist Assistant client is installed in C:\Program Files\Softgenetics\Geneticist Assistant and you cannot change this location. The Apache HTTP server is installed in the directory C:\Program Files (x86)\Apache2.2_forGA and you cannot change this location. The Python programming language is installed to a directory on your hard drive with the version number embedded. For example, version 2.7 of Python is installed in C:\Python27.

The Geneticist Assistant executable is configured to look for any available local port during installation; however, if an error message is displayed that the Apache service could not be started, you might have a conflict with an application that is already using the selected port. Contact SoftGenetics for assistance.

Starting and Logging into Geneticist Assistant

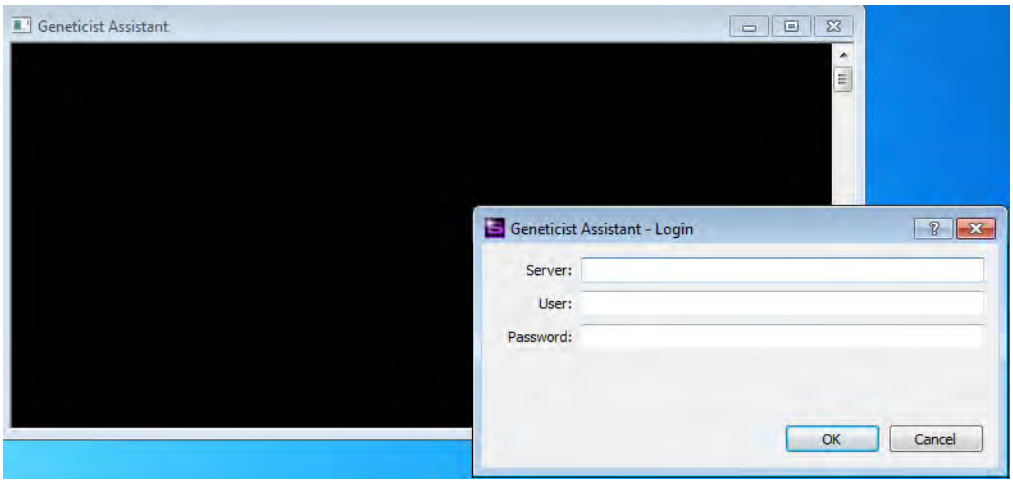
After you install Geneticist Assistant, a shortcut icon for the application is placed on the desktop. An option for the application is also available from the Start menu. After Geneticist Assistant is installed and you open Geneticist Assistant for the first time, you must specify the URL for the database server.

Figure 1-4: Geneticist Assistant desktop icon

1. Double-click the desktop icon to launch the application, or select the option from the Start menu (Start > All Programs > SoftGenetics > Geneticist Assistant).

   The Geneticist Assistant Login dialog box and a Geneticist Assistant Debug window open.

Figure 1-5: Geneticist Assistant Login dialog box and Debug window



2. If this is the *first* time that you are logging into Geneticist Assistant, then do the following; otherwise, go directly to Step 3.

   - In the Server field, enter the URL for the Geneticist Assistant database server:

Installation Type	Description
Local	The database server is installed on the local computer. The URL for this is https://localhost.
Intranet	The database server is installed on a different computer that is on your network. The URL must include the computer name or IP address.
Internet	The database server is installed on a computer that has Internet access. The computer can be outside your local network. The URL must include the computer name or IP address. **Note:** Geneticist Assistant uses SSL encryption to ensure the security of the data over the Internet.

   - In the Username field, enter your username, and then in the Password field, enter your password, and then click OK.

For subsequent Geneticist Assistant sessions, the Username field is automatically populated with the username for the user who last logged in, and the Server field is automatically populated with the URL for the database server.

3. If needed, in the Username field, enter your username, and then in the Password field, enter your password, and then click OK. One of the following happens:

   - If your login is successful, then the Geneticist Assistant Login dialog box closes, "Success" is displayed in the Debug window, and the Geneticist Assistant main window opens. The Debug window remains open.
   - If your login is not successful, then the reason for the failure is displayed in the Debug window. If you are unable to resolve the login issues based on the messages in the Debug window, contact SoftGenetics for assistance.

The Debug window displays the results after each step that you carry out in Geneticist Assistant. You must leave this window open for the duration of your session. If you close it, then you also close Geneticist Assistant.

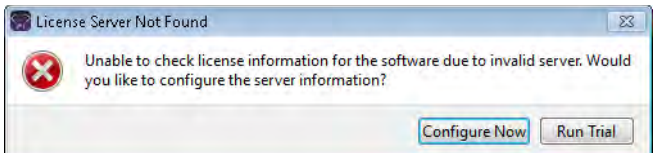
4. Continue to one of the following:

   - If you are running a trial version of Geneticist Assistant, continue to "To run a trial version of Geneticist Assistant" on page 19.
   - If you are running a purchased version of Geneticist Assistant, continue to "To run a purchased version of Geneticist Assistant" on page 20.

To run a trial version of Geneticist Assistant

After you log into Geneticist Assistant, if you are running a trial version of Geneticist Assistant, then a License Server Not Found message *always* opens.

Figure 1-6: License Server Not Found message

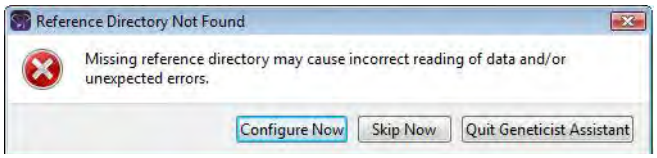


1. Click Run Trial.

   The License Server Not found message closes. Two results are possible.

   - If this is the first time that you are running the trial version, then a Reference Directory Not Found message opens. You must configure your reference directory before you can use the software. Continue to Step 2.

Figure 1-7: Reference Directory Not Found message



   - If this is *not* the first time that you are running the trial version, and the reference directory has already been configured, then the Geneticist Assistant main window opens. Continue to "The Geneticist Assistant Main Window" on page 23.

2. Do one of the following:

   - To configure the reference directory now, click Configure Now to open the Settings dialog box, and then continue to "To configure the reference directory" on page 22.
   - To configure the reference directory at a later date, click Skip Now.

     The Geneticist Assistant main window opens.

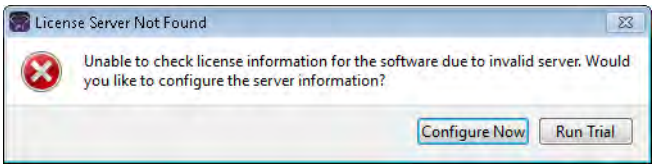
3. Continue to "The Geneticist Assistant Main Window" on page 23.

For detailed information about configuring the required reference directory, see "To configure the reference directory" on page 46.

To run a purchased version of Geneticist Assistant

If you are running a purchased version of Geneticist Assistant, then the first time that you run the application, a License Server Not Found message opens. You must configure Geneticist Assistant to connect to the SoftGenetics license server. After you configure the license server, this message does not open again.

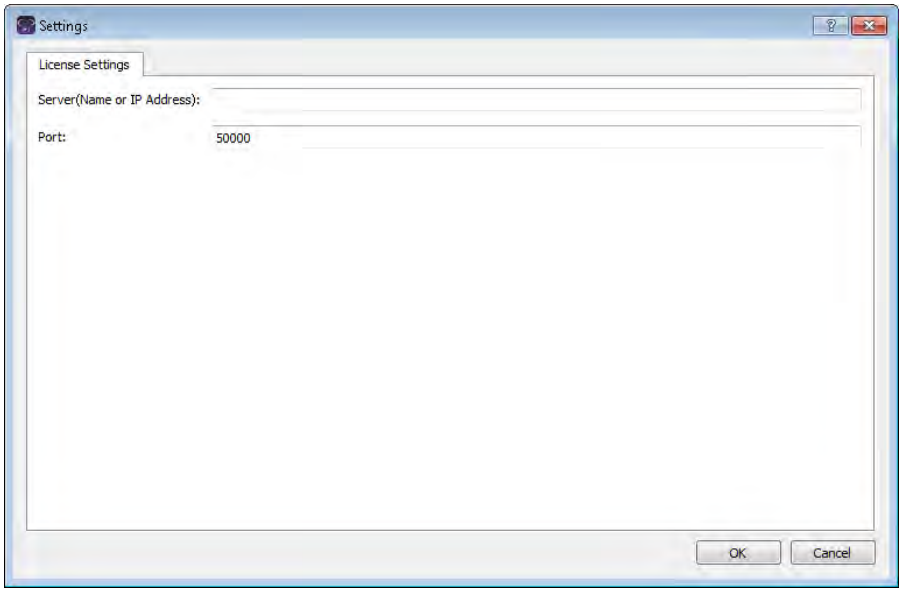
Figure 1-8: License Server Not Found message



1. Click Configure Now.

   The Settings dialog box opens. The License Settings tab is the only tab on the dialog box.

Figure 1-9: Settings dialog box



2. In the server name, enter the name for the server, or its IP address.

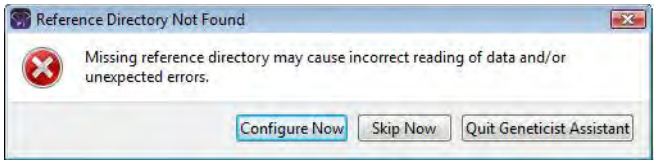
If the SoftGenetics license server is running on the same computer on which Geneticist Assistant is running, then you must enter 127.0.0.1.

3. Leave the Port value set to the default value of 50000.

4. Click OK.

   The Settings dialog box closes, and a Reference Directory Not Found message opens. You must configure your reference directory before you can use the software.

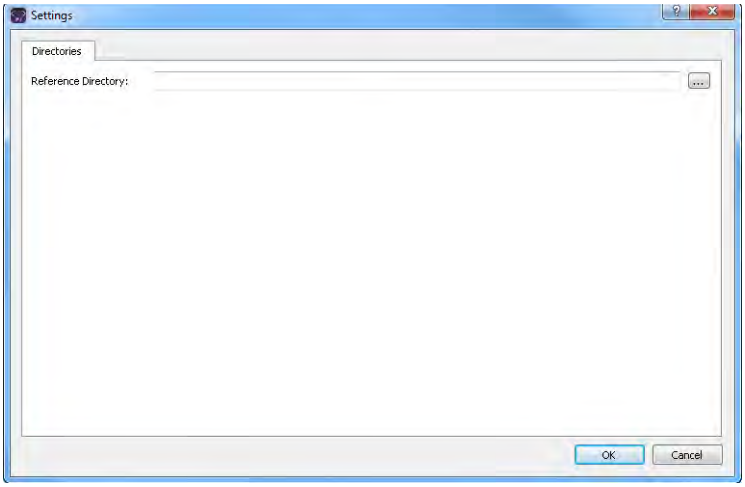
Figure 1-10: Reference Directory Not Found message



5. Do one of the following:

   - To configure the reference directory now, click Configure Now to open the Settings dialog box, and then continue to "To configure the reference directory" on page 22.

Figure 1-11: Settings dialog box, Directories tab



   - To configure the reference directory at a later date, click Skip Now.

     The Reference Directory Not Found message closes and the Geneticist Assistant main window opens. Continue to "The Geneticist Assistant Main Window" on page 23.

For detailed information about configuring the required reference directory at a later date, see "Specifying your Geneticist Assistant Settings" on page 45.

To configure the reference directory

Before you run Geneticist Assistant for the first time, you must configure the reference directory. The *reference directory* contains the information for the reference that was used for aligning the data and/or detecting variants. References files are stored in their own folders in this directory. For example, if you are studying variants that are associated with the human genome, then you might create a Human folder in the Reference directory, such as C:\References\Human 37.

After you specify the location for the reference directory, and submit data to the Geneticist Assistant database, you cannot change its location.

1. Create your reference directory in a location that meets your business needs.

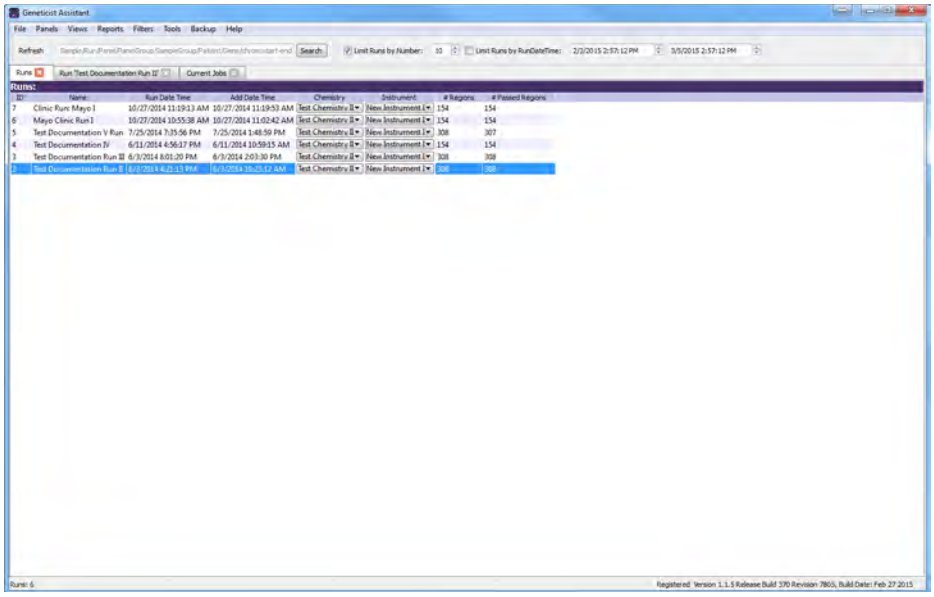
Although not required, SoftGenetics recommends that you create this directory on the same computer and drive on which you installed the Geneticist Assistant client.

2. Click the Browse button  next to the Reference Directory field, and then browse to and select the Reference directory.
3. Click OK to close the Settings dialog box and return to the Geneticist Assistant main window.
4. Continue to "The Geneticist Assistant Main Window" on page 23.

The Geneticist Assistant Main Window

The Geneticist Assistant main window opens automatically after you successfully log into the Geneticist Assistant application.

Figure 1-12: Geneticist Assistant main window



The Geneticist Assistant main window is your starting point for the Geneticist Assistant application. The window provides quick access to all of the Geneticist Assistant functions. From top to bottom, the Geneticist Assistant main window has five major components—the title bar, the main menu, the toolbar, the tabs display area, and the status bar.

Title bar

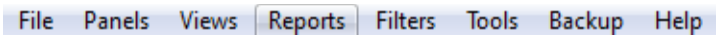
The name “Geneticist Assistant” is displayed in the title bar at the top of the Geneticist Assistant main window.

Figure 1-13: Geneticist Assistant title bar

Main menu

The main menu is set up in a standard Windows menu format with menu commands grouped into menus (File, Panels, References, Views, Reports, Filters, Tools, Backup, and Help) across the menu bar.

Figure 1-14: Geneticist Assistant main menu

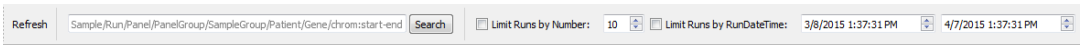


Toolbar

The Geneticist Assistant main window toolbar is located below the main menu. It always contains a Refresh option and a Search feature. If the Runs tab or the Patients tab is open, it also contains options for limiting the display on the tab.

- You use the Refresh option to manually refresh a tab display after the tab opens in the main window and data is loaded, or after adding, modifying, and/or deleting data on a tab. See "Main window tabs" below.
- You use the Search feature to search for samples, runs, panels, panel groups, sample groups, genes, and variants in your Geneticist Assistant database. See "Searching the Geneticist Assistant Database" on page 36.

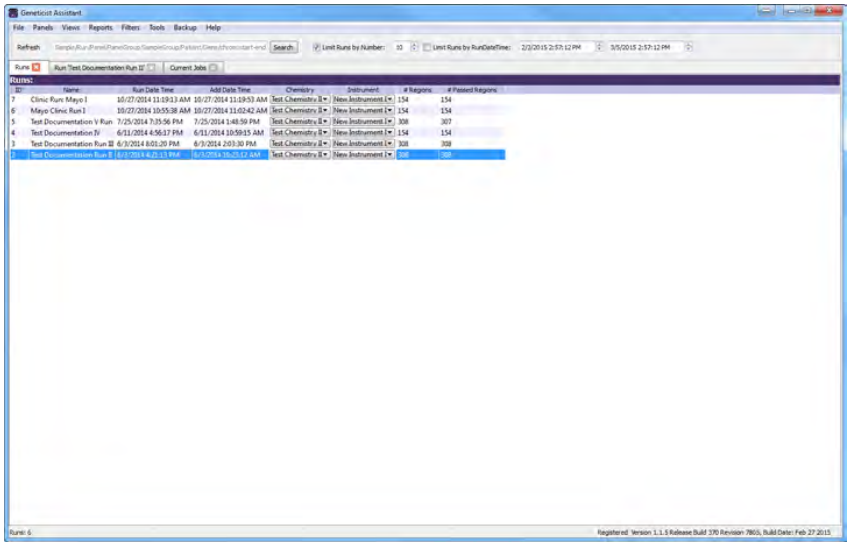
Figure 1-15: Geneticist Assistant main window toolbar



Main window tabs

When the Geneticist Assistant main window first opens, two tabs— Runs and Current Jobs—are displayed in the window. The Runs tab is the open tab. A *run* refers to loading a BAM or DAT file *and* a VCF file or just a VCF file into Geneticist Assistant for the purposes of importing a list of variants into the Geneticist Assistant database and/or reviewing a list of variants. The Runs tab has a single pane—the Runs pane.

Figure 1-16: Geneticist Assistant main window, Runs tab with Runs pane



- If you are an Administrator user, then the Runs pane can display all the runs submitted by all users to your Geneticist Assistant database. By default, when the tab first opens, only up to the 10 most recently submitted runs are displayed and the runs are sorted based on the Run ID. You can limit the runs that are displayed by number and/or by a time and date range and you can also change the sort order of the data.

- If you are a standard user, then the Runs pane can display all the runs that you have submitted to your Geneticist Assistant database as well as all the runs that have been submitted by other users and for which you have been granted at least the Can Read permission. By default, when the tab first opens, only up to the 10 most recently added runs that fit one of these two criteria are displayed and the runs are sorted based on the Run ID. You can limit the runs that are displayed by number and/or by a time and date range and you can also change the sort order of the data.

If you close the Runs tab, you can open it again from the Geneticist Assistant main menu: File > Runs.

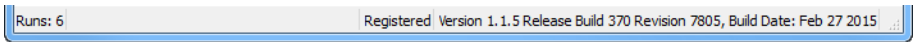
The Current Jobs tab displays the progress of a run. The tab opens automatically after a run is started. After the run is successfully completed (a BAM or DAT file *and* a VCF file are loaded and read, or a VCF file is loaded and read), the Current Jobs tab is cleared and remains blank. Other tabs are available in the display area of the Geneticist Assistant main window depending on the actions that you carry out.

With the exception of the Current Jobs tab, every tab in Geneticist Assistant has at least one pane. The different panes display different information. Data is displayed in columns in the tab panes. A variety of options are available for setting preferences for the tab display and the data column display. See "Setting Display Preferences in the Geneticist Assistant Main Window."

Status bar

The Status bar is displayed at the bottom of the Geneticist Assistant main window. Summary information that is tab-specific is displayed in the left corner of the status bar. Release information (Version, Release Build, Revision, and Build Date) is displayed in the right corner of the status bar. This release information is identical to the release information that is displayed from the Help > About menu option.

Figure 1-17: Geneticist Assistant main window, Status bar



Setting Display Preferences in the Geneticist Assistant Main Window

With the exception of the Current Jobs tab, every tab in Geneticist Assistant has at least one pane. The different panes display different information. Data is displayed in columns in the tab panes. A variety of options are available for setting preferences for the tab and tab pane displays and the data column display. You can:

- Navigate multiple open tabs/minimized window. See “Navigate multiple open tabs/ Navigate tabs with a minimized main window” on page 26.
- Close multiple tabs in a single step. See “Close multiple tabs in a single step” on page 27.
- Show/hide the toolbar for a tab. See “Show/Hide the toolbar for a tab” on page 27.
- Show/hide a tab pane. See “Show/hide a tab pane” on page 27.
- Filter the data that is displayed in a pane. See “Filter the data that is displayed in a pane” on page 27.
- Work with data columns in a tab pane. See “Work with columns in a tab pane” on page 30.
- Change the widths of a data columns. See “Change the widths of data columns” on page 35.
- Rearrange the order of data columns. See “Rearrange the order of data columns” on page 35.
- Change the sort order of data. See “Change the sort order of data” on page 35.
- Rename column headers. See “Rename column headers” on page 35.
- Select the data that is to be included in a custom report. See “Select data to be included in a custom report” on page 35.

Any preferences that you set for a tab display remain in effect regardless if you log out of and then log back into Geneticist Assistant. You can set the preferences for the Variants Table pane on the Coverage QC tab or the Variants pane on the Sample tab, and then apply these same settings to the appropriate pane on the other tab. You can also return the tab displays to their default displays (all tab panes shown, all data columns shown, and so on) in a single step. See “To apply shared preferences or to clear all tab preferences” on page 51.

Navigate multiple open tabs/Navigate tabs with a minimized main window

If you open multiple tabs, or if you have reduced the size of the main window, then you might not be able to see all the tabs at the top of the main window, and the display options at the top of the Runs tab or the Patients tab might also be hidden. You can use the Back and Next arrows  that are displayed in the top right corner of the main window to move through all open tabs and change the display focus, and/or view the hidden display options.

Close multiple tabs in a single step

If you have opened multiple tabs, then you close each tab individually, or you can close all tabs in a single step, or you can close all tabs other than a selected tab in a single step. To close all tabs in a single step, right-click the title bar for any of the opened tabs, and on the context menu that opens, click Close All. To close all tabs other than a selected tab, right-click the title bar of the tab that is to remain open, and on the context menu that opens, click Close All Except This.

Show/Hide the toolbar for a tab

The Views menu contains an option to show or hide the toolbar for a tab. By default, when a tab first opens, the toolbar is displayed for a tab. To hide the toolbar for the tab, you can clear the check that is displayed next to Tool Bar option. To show the toolbar for a tab, select Tool Bar again.

Show/hide a tab pane

By default, when a tab first opens, all available tab panes are displayed. On the Views menu, you can clear the check that is displayed next to a pane name to hide the entire pane from view. To show the pane, select the pane name again. An option to Show All Views is available on the Views menu for some tabs. If any of the panes on a tab are hidden, then you can select this option to show all panes again in a single step.

Filter the data that is displayed in a pane

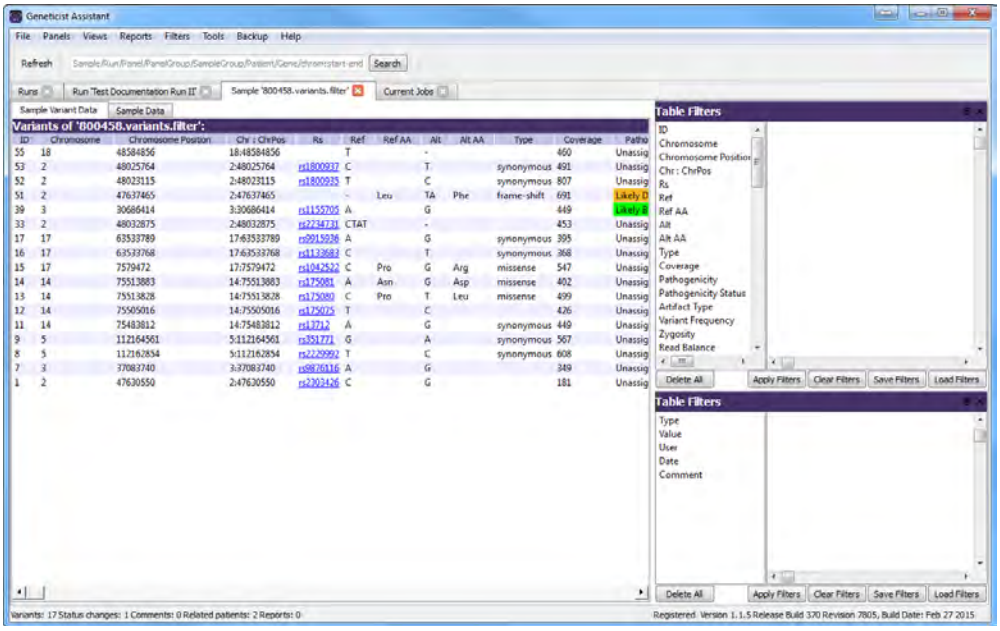
By default, when a tab first opens, all data in all available panes is displayed. A Filters table displays all the data (fields) for a selected tab on a pane. You specify the filters in a Filters table to limit the display in a tab pane to only the data (fields) that meet specific criteria. You can specify the filters for the data that is displayed in a tab pane on a one time only basis, or you can save the filters to a text file, and then load the file to filter the data that is displayed in the tab pane anytime as needed.

1. Open the appropriate tab.
2. On the Geneticist Assistant main menu, click Filters > <Tab Pane>. For example, if you opened the Filters pane with the Sample tab open, then the menu options would be Filters > Sample's Variants, Filters > Sample's Status Changes, and so on.

   The Filters table opens to the right of the tab. The table lists all the fields for the selected tab pane. See Figure 1-18 on page 28.
3. Repeat Step 2 as many times as needed to open multiple Filters tables for a tab.

The remainder of this procedure describes how to set up new filters to filter the data that is displayed in a tab pane. Optionally, you can click Load Filters to open the Load Filter Template dialog box and select a filter template to filter the data display based on the saved filter settings in the template.

Figure 1-18: Filters tables opened for a tab

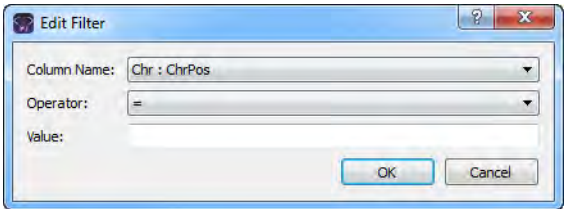


4. Click and drag the field by which to filter the data *from* the list of available fields *to* the blank pane that is to the right of the list.

   An Edit Filter dialog box opens. The name of the selected field is displayed in the Column Name field on the dialog box.

The Column Name is a dropdown list that contains all the available fields for the opened tab. You can always select a different field on this list before continuing to Step 5.

Figure 1-19: Edit Filter dialog box



5. On the Operator dropdown list, select the operator for the filter.

The operators that are available on the Operator dropdown list are specific to the type of field that is selected - text, numeric, and so on. Not all operators are available for all field types.

Filter Operator	Description
=	Equals. Must be an exact match to return data.

Filter Operator	Description
≠	Does not equal. Only data that does not match the value specified is returned.
>	Greater than. Only data that exceeds the value specified is returned.
<	Less than. Only data that is less than the value specified is returned.
>=	Greater than or equal to. Only data that is an exact match for or exceeds the value specified is returned.
<=	Less than or equals to. Only data that is an exact match for or is less than the value specified is returned.
contains	Returns data that not only contains the value specified, but also might contain other information. The search string is not case-sensitive.
does not contain	Returns only the data that does not contain the value specified. The search string is not case-sensitive.
On	Only data with a date stamp that is the same as the specified date is returned. Identical to the = operator, but used specifically for dates.
Earlier than	Only data with a date stamp that falls before the specified date is returned. Identical to the < operator, but used specifically for dates.
Earlier than or on	Only data with a date stamp that is the same as the specified date or that falls before the specified date is returned. Identical to the <= operator, but used specifically for dates.
Later than	Only data with a date stamp that falls after the specified date is returned. Identical to the > operator, but used specifically for dates.
Later than or on	Only data with a date stamp that is the same as the specified date or that falls after the specified date is returned. Identical to the >= operator, but used specifically for dates.

6. Enter the filter value.
7. Click OK.

   The Edit Filter dialog box closes. The selected field, its operator, and its value are displayed in the panel that is next to the list of available filter fields.

8. Repeat Step 4 through Step 7 for each field by which the data is to be filtered. When you filter the data by multiple fields, two options are possible:

   - You can drag separate additional fields for filtering into the blank pane to apply filters on an “or” basis. For example, for the Sample's Variants sub-tab on the Sample tab, you can add filters for Coverage >= 300 and a filter for Gene = MSH6. The results are filtered to show those variants that have a coverage greater than or equal to 300 *or* variants that are found in the MSH6 gene.
   - You can drag additional fields for filtering on top of an existing filter field to apply the filters on an “and” basis. For example, for the Sample's Variants sub-tab on the Sample tab, you can add the filter for Coverage >= 300 and then drag the filter for Gene = MSH6 onto the Coverage filter. The results are filtered to show only those variants that have a coverage greater than or equal to 300 *and* that are found in the MSH6 gene.

9. Do any of the following as needed:

    - Click Apply Filters to immediately apply the filters and view the results onscreen. ("*Filters Applied" is displayed in the title of the affected tab panes.)

If the results are not as expected, this allows you to make the necessary adjustments to the filter before saving it.

    - Click Clear Filters to reset the display for all panes on a tab in a single step. The filters remain displayed in the Filter table.
    - Right-click the filter in the filter pane, and on the context menu that opens, click Edit to open the Edit Filter dialog box, and edit the filter (Column, Operator, and/or Value) as needed, and then click OK to close the dialog box and save the edited filters. You can then click Apply Filters to immediately apply the edited filters and view the updated results onscreen.
    - Right-click the filter in the filter pane, and on the context menu that opens, click Remove to delete the filter. You can then click Apply Filters to immediately remove the filters from the display and view the updated results onscreen.
    - Click Save Filters to open the Save Filter Template dialog box and save your filter settings to a file. You can use this saved Filter Settings file to filter the data for another project based on the settings in the file.
    - Click Delete Filters to reset the display for all panes on a tab in a single step and delete all the filters from the Filter table.

10. After you are done filtering the data, you can click Close (x) in the upper right corner of a Filter table to close it.

Work with columns in a tab pane

When working with columns in a tab pane, you can do any or all the following:

- Show/hide columns dynamically in a tab pane. See "To show/hide columns in a tab pane" below.
- Create a pane configuration. See "To create/modify a pane configuration" on page 32.
- Load a pane configuration. See "To load a pane configuration" on page 33.
- Delete a pane configuration. See "To delete a pane configuration" on page 34.

To show/hide columns in a tab pane

The Select/deselect/restore all option on the Select Columns dialog box is a tri-state checkbox. When the dialog box first opens, for a tab pane, all the columns are selected for display, and the Select/deselect/restore option is also selected. If you click it once, then all data columns are hidden from view in the pane in a single step, and the Select/deselect/

restore option is cleared. If you click this option a second time, then the last specified settings (that is, which columns were shown and which columns were hidden) are restored, and a completely filled checkbox is displayed for the Select/deselect/restore option. If you click tis option a third time, then all the columns are again selected for display, and the Select/deselect/restore option is also selected again.

1. Right-click the tab pane anywhere, and on the context menu that opens, click Load Columns.

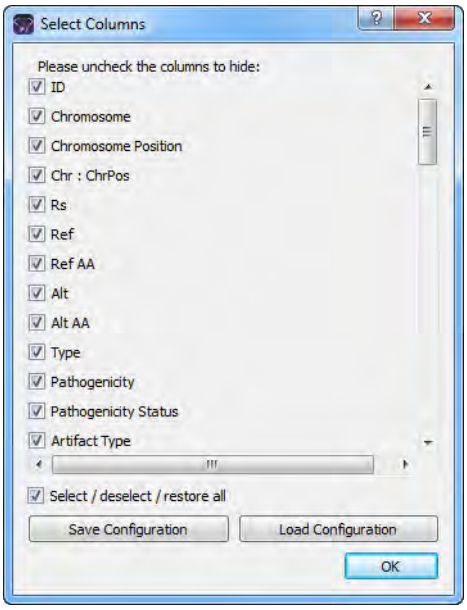
   A sub-menu opens that displays both a Manually option and all the publicly available configurations for the pane. The name of the user who created each configuration is also displayed.

2. Do one of the following:

   - Select one of the public configurations to display the columns as specified in the configuration.
   - Click Manually.

     The Select Columns dialog box opens. By default, when the dialog box first opens for a tab pane, all the columns are selected for display, and Select/deselect/restore all is also selected. Continue to Step 3.

Figure 1-20: Select Columns dialog box



3. Clear the options for the columns that are to be hidden from view in the pane, and leave the options for the columns that are to be shown selected.

   The pane display is dynamically updated with your selections.

4. Click Close (x) to close the dialog box.

   The column preferences remain in effect even if you log out of and then log back into Geneticist Assistant. To restore the last specified settings, or display all columns in a single step, click the Select/deselect/restore all option as needed.

To create/modify a pane configuration

When you create and save a configuration for a tab pane, the configuration is available and applicable only for this specific tab pane. You can modify only the configurations that you have created.

1. Right-click the tab pane anywhere, and on the context menu that opens, click Load Columns.

   A sub-menu opens that displays both a Manually option and all the publicly available configurations for the pane. The name of the user who created each configuration is also displayed.

2. Do one of the following:

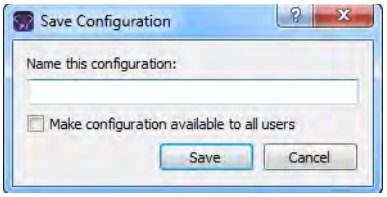
Option	Description
To create a new pane configuration by modifying an existing configuration	1. Click Manually. The Select Columns dialog box opens. See Figure 1-20 on page 31. 2. Click Load Configuration. The Load Configuration dialog box opens. The dialog box displays all your private and public configurations as well as all the configurations that other users have made Public. *Figure 1-21: Load Configuration dialog box*  3. Select the configuration that is to be modified. The pane display is dynamically updated according to the loaded configuration 4. Right-click the tab pane anywhere, and on the context menu that opens, click Load Columns > Manually. The Select Columns dialog box opens again. The settings on the dialog box reflect the loaded configuration. 5. Continue to Step 3.

Option	Description
To create an entirely new pane configuration	1. Click Manually. The Select Columns dialog box opens. By default, when the dialog box first opens for a tab pane, all the columns are selected for display, and Select/deselect/restore all is also selected. See Figure 1-20 on page 31. 2. Continue to Step 3.

3. Clear the options for the columns that are to be hidden from view in the pane, and select the options for the columns that are to be displayed.
4. Click Save Configuration.

   The Save Configuration dialog box opens.

Figure 1-22: Save Configuration dialog box



5. Enter a name for the configuration.

If you are modifying a configuration, then you must save it with exactly the same name as the original configuration.

6. If this configuration is to be available to all others of this Geneticist Assistant instance (a *public* configuration), then select Make configuration available to all users; otherwise, to make the configuration available only to you (a *private* configuration), leave this option blank.
7. Click Save.

   The Save Configuration dialog box closes. The Select Columns dialog box remains open.
8. Click Close (x) to close the Select Columns dialog box.

To load a pane configuration

1. Right-click the tab pane anywhere, and on the context menu that opens, click Load Columns.

   A sub-menu opens that displays both a Manually option and all the publicly available configurations for the pane. The name of the user who created each configuration is also displayed.

2. Select the configuration that you are loading.

   The Select Columns dialog box close. The pane display is updated dynamically after you load the configuration.

To delete a pane configuration

You can delete only those pane configurations that you have created.

1. Right-click the tab pane anywhere, and on the context menu that opens, click Load Columns.

   A sub-menu opens that displays both a Manually option and all the publicly available configurations for the pane. The name of the user who created each configuration is also displayed.

2. Click Manually.

   The Select Columns dialog box opens.See Figure 1-20 on page 31.

3. Click Load Configuration.

   The Load Configuration dialog box. The dialog box displays all your private and public configurations as well as all the configurations that other users have made public.

Figure 1-23: Load Configuration dialog box

Although all these configurations are displayed, you can delete only those configurations that you have created.

4. Right-click the configuration that you are deleting, and on the context menu that opens, click Delete Column Configuration.

   The Column Configuration is deleted.

If you select a configuration that you did not create, then the Delete Column Configuration option is not available on the context menu.

5. Click Close (x) to close the Load Configuration dialog box.
6. Click Close (x) to close the Select Columns dialog box.

Change the widths of data columns

To change the width of a data column, rest the mouse pointer on the right side of a column heading until the pointer changes to a double-headed arrow, and then drag the boundary until the column is the width that you want.

Rearrange the order of data columns

You can rearrange the order of the data columns in a tab pane by clicking and holding the mouse pointer on a column header, and then dragging the column to a new location.

Change the sort order of data

When a tab first opens, data is sorted in ascending order based on the IDs of the data. To change the sort order of the data in a column, click a column header to sort the display based on the descending order of data in the column. A Sort Order icon is displayed in the column header. You can click this Sort Order icon to toggle between an ascending sort and a descending sort of the data based on the column heading.

Rename column headers

Every data column is titled with a default column header. To rename a column header to better suit your working needs, right-click the column header, and on the context menu that opens, click Rename Header. The column header is selected, and you can then modify the column header as needed.

Select data to be included in a custom report

You can include just specific data in a custom report, or you can include all data in a pane in a custom report.

- To include just specific data, right-click the data (press and hold the CTRL key to select multiple rows of data), and on the context menu that opens, select Add Selected to Custom Report. *All* data in all columns that are currently shown in the selected rows is included in the custom report. Data in hidden columns is not included in the report.
- To include all data that is currently shown in a tab pane in a custom report, right-click anywhere in the pane and on the context menu that opens, click Add All to Custom Report. *All* data in all columns that are currently shown in the tab pane is included in the custom report. Data in hidden columns is not included in the report.

For detailed information about generating a custom report after you have selected the necessary data, see *"Generating a Custom Report" on page 209.*

Searching the Geneticist Assistant Database

You use the Search feature that is available on the Geneticist Assistant main window toolbar to search for samples, runs, panels, panel groups, sample groups, genes, and variants in your Geneticist Assistant database.

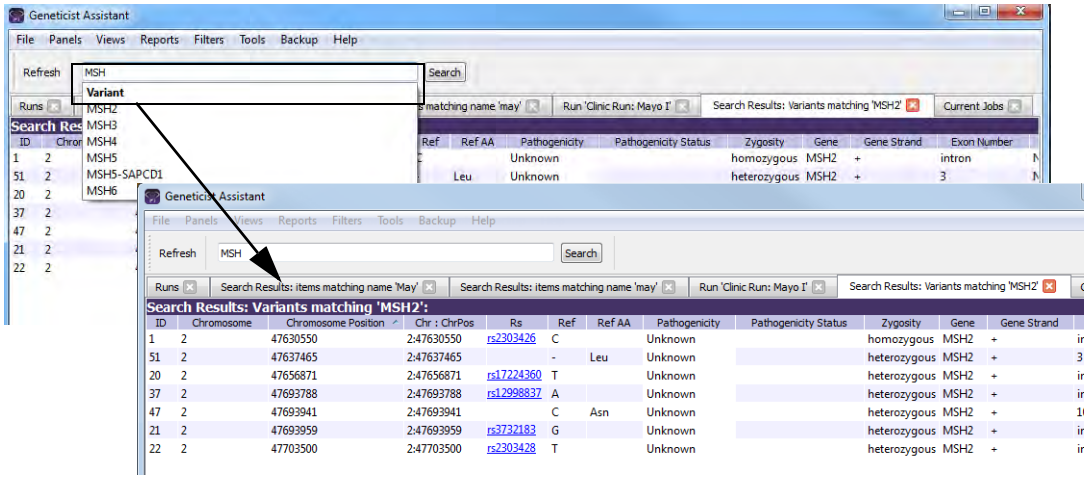
Figure 1-24: Geneticist Assistant main window toolbar

Important points to note about a search are the following:

- As you enter a search string, the list of search results that match the string is dynamically updated, and the type of search result that matches the string is indicated in bold above the list (**Run**, **Variant,** and so on).
- The search for any item is not case-sensitive, and the search string can be found anywhere in the search results. For example, if you enter “Mayo” as the search string, then the search results can include a run named “Mayo Clinic” or a run named “Clinic Run:Mayo.”
- If you search for a variant, the search is not carried out relative to any sample, and multiple search options are available.
  - You can use the variant description in the format of chrom:start position:ref:alt. You must separate multiple variant search strings with a comma (,) or a semi-colon (;).
  - You can search for a single position on a single chromosome (for example, 1:123) or for a range of positions on a single chromosome (for example, 1:1-1000).
  - You can search by the partial or complete gene name; for example, MSH or MSH2, respectively.

Search results for any entity in your Geneticist Assistant database are displayed on a Search Results tab.

Figure 1-25: Search feature and results example



You can double-click any search result on a Search Results tab to “drill down” into the search result, and review the associated data. For example, you can:

- Double-click a Run search result to open the associated Run tab.
- Double-click a Gene search result to open the list of variants for the gene, and then double-click a variant in this list to open the associated Variant tab.

You can also right-click any search result to open a context menu that has options that are specific for the search result.

Chapter 1
Getting Started with Geneticist Assistant

Chapter 2
Geneticist Assistant Setup and Maintenance

Before you use Geneticist Assistant for the first time, you must add and/or modify the necessary objects and specify the settings for your installation. For data protection, you can manually back up the Geneticist Assistant database at any time of your choosing.

This chapter covers the following topics:

Chapter 2
Geneticist Assistant Setup and Maintenance

Managing Objects

An *object* in Geneticist Assistant holds a value or variable that can be applied for carrying out a run in Geneticist Assistant, or for processing and interpreting the resulting run data. Managing objects in Geneticist Assistant consists of adding and editing the values for these objects as well as deleting values that are no longer valid or required. Ten objects are available in Geneticist Assistant—Chemistry, Instrument, Pathogenicity, Pathogenicity Status, Sample Status, Panel Group, Coverage Region Status, Sample Status, Run Status, and Artifact Type. Geneticist Assistant is installed with default values for Pathogenicity, Pathogenicity Status, Sample Status, Coverage Region Status, Run Status, and Artifact Type. You have the option of adding custom values for these objects, and then selecting a custom value in lieu of a default value. You must manually add values for all other objects.

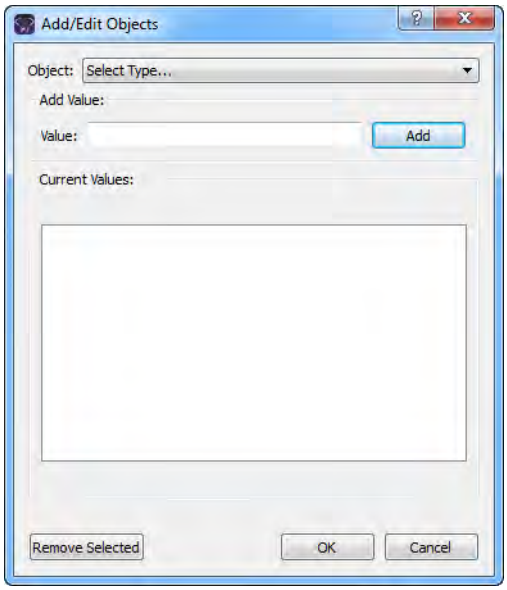
Object	Description
Chemistry	Refers to the chemistry that was used to prepare the samples for sequencing.
Instrument	Refers to the instrument that was used to sequence the samples.
Pathogenicity	The pathogenicity for a selected variant. Default values are Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign.
Pathogenicity Status	Indicates the status for changing the pathogenicity for a selected variant. Default values are Pending and Confirmed.
Sample Status	The status for a sample. Default values are New, QC Passed, Reviewed, and Complete.
Panel Group	Separately tracks the pathogenicities of variants that might be found in multiple panels. For example, the pathogenicity of a selected variant is set to Deleterious for multiple cancer panels; however, the pathogenicity can be set to Benign for a cardiac panel. You can create a group for each panel, and then assign the appropriate panels to each group.
Coverage Region Status	The Coverage Regions tab (via the Run tab) or the Coverage Regions sub-tab on the Coverage QC tab (via the Run tab) lists the coverage information for every region in your panel as well as a status for each region. Default values are Passed or Failed to indicate whether a region passed or failed the coverage settings that were set for the run. **Note:** See "Coverage Regions sub-tab (Coverage QC tab)" on page 146.
Sample Group	Used for a group of samples that are to be processed repeatedly over a period of time, for example, a group of control samples for which the coverage or variant frequency is to be tracked.
Run Status	The status for a run. Default values are New, QC Passed, Reviewed, and Complete.
Artifact Type	The artifact type for the sample variant, for example, a Sequencing artifact. A single default value, None, is supplied for artifact type.

To add a value for an object

1. On the Geneticist Assistant main menu, click File > Add/Edit Objects.

   The Add/Edit Objects dialog box opens.

Figure 2-1: Add/Edit Objects dialog box



2. On the Object dropdown list, select the object for which you are adding a value.

   If values have already been added for the selected object, or the selected object has default values, then these values are displayed in the Current Values pane.

3. In the Value field, enter the value for the object, and then click Add.

The value can have a maximum of 255 characters. Spaces and special characters are allowed.

   The newly added value is displayed in the Current Values pane.

4. If you added a new pathogenicity value, then to reorder the lists of displayed values, select a value in the Current Values pane, and then click Move Up or Move Down as needed.

This is a required step to correctly order the pathogenicity values from least severe to most severe.

5. Click OK.

   The message “Saving new objects” is displayed on the Add/Edit Objects dialog box. The dialog box closes after the value is added successfully.

If you add a value for pathogenicity, you can also specify a highlighting color for variants of this type in all the variant panes and sub-tabs. See "To specify the highlighting colors for variants of a pathogenicity" on page 48.

To edit an object

When you edit an object, you edit the object value. Any previous runs that use the edited object value are not affected. The "old" (previous) value remains displayed in Geneticist Assistant on the Runs tab. Going forward, only the "new" value is available for selection for new runs.

1. On the Geneticist Assistant main menu, click File > Add/Edit Objects.

   The Add/Edit Objects dialog box opens. See Figure 2-1 on page 42.

2. On the Object dropdown list, select the object for which you are editing a value.

   The currently available values for the selected object are displayed in the Current Values pane.

3. In the Current Values pane, double-click the object value that you are editing.

   The value is highlighted in a blue rectangle.

4. Edit the value as needed.

The value can have a maximum of 255 characters. Spaces and special characters are allowed.

5. Click OK.

   The message "Saving new objects" is displayed on the Add/Edit Objects dialog box. The dialog box closes after the value is edited successfully.

To delete a value from an object

If you delete a value for an object that has been previously used in a run, then the run is also deleted in its entirety from the Geneticist Assistant database. Be very sure that you want to delete a value from an object before doing so.

1. On the Geneticist Assistant main menu, click File > Add/Edit Objects.

   The Add/Edit Objects dialog box opens. See Figure 2-1 on page 42.

2. On the Object dropdown list, select the object from which you are deleting values.

   The currently available values for the selected object are displayed in the Current Values pane.

3. In the Current Objects pane, click the object value that you are deleting, and then click Remove Selected.

   The value is deleted immediately. The Add/Edit Objects dialog box remains open.

4. Click OK.

   The Add/Edit Objects dialog box closes.

Specifying your Geneticist Assistant Settings

Before you run Geneticist Assistant for the first time, you must create your reference directory. The *reference directory* contains the information for the reference that was used for aligning the data and/or detecting variants. References files are stored in their own folders in this directory. For example, if you are studying variants that are associated with the human genome, then you might create a Human folder in the Reference directory, such as C:\References\Human 37. When specifying the settings for your Geneticist Assistant installation, at a minimum, you *must*:

- Create your reference directory and specify its location. See "To configure the reference directory" on page 46.
- Specify the global Quality Control (coverage) settings for your data. See "To specify the global Quality Control settings" on page 47.

Optionally, you can also:

- Specify the colors for highlighting variants of a specific pathogenicity in all the variant panes and variant sub-tabs. See "To specify the highlighting colors for variants of a pathogenicity" on page 48.
- Specify the same column display preferences for variants in all the variant panes and variant sub-tabs (with the exception of the Sample Group columns on the Sample Variant Data sub-tab for the Sample tab and the Variant pane on the Variant tab), and/or clear the preferred settings for all the data tabs. See "To apply shared preferences or to clear all tab preferences" on page 51.
- Specify the preferred settings for variants, which determines how the information is displayed for variants in all the variant panes and variant sub-tabs, and/or clear the preferred settings for all the data tabs. See "To specify variant preferences" on page 53.
- Specify your Alamut settings. See "To specify Alamut settings" on page 54.
- Specify your HGMD settings. See "To specify HGMD settings" on page 56.
- Specify the requirements for all user passwords for Geneticist Assistant. See "To specify the Geneticist Assistant password requirements" on page 57.
- Specify your OMIM settings. See "To specify the OMIM settings" on page 58.
- Modify the SoftGenetics license server settings. See "To modify the SoftGenetics license server settings" on page 59.

To configure the reference directory

You carry out the following procedure if you did not configure the reference directory the first time that you logged into Geneticist Assistant, or if you want to change its location.

If you have previously specified the location for the reference directory and submitted data to the Geneticist Assistant database, you cannot change its location.

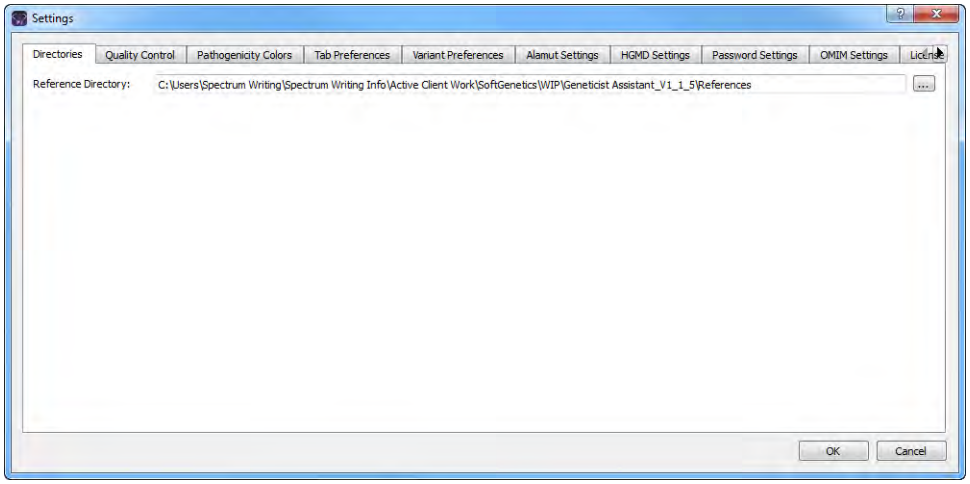
1. Create your reference directory in a location that meets your business needs.

Although not required, SoftGenetics recommends that you create the reference directory on the same computer and drive on which you installed the Geneticist Assistant client.

2. On the Geneticist Assistant main menu, click File > Settings.

   The Settings dialog box opens. By default, the Directories tab is the open tab.

Figure 2-2: Settings dialog box, Directories tab



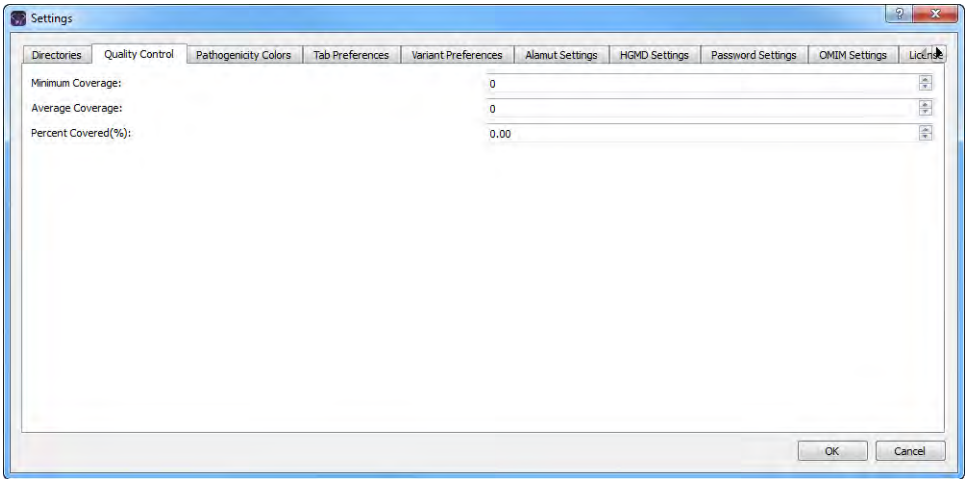
3. Click the Browse button ... next to the Reference Directory field, and then browse to and select the Reference directory.
4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - “To specify the global Quality Control settings” on page 47.
   - “To specify the highlighting colors for variants of a pathogenicity” on page 48.
   - “To apply shared preferences or to clear all tab preferences” on page 51.
   - “To specify variant preferences” on page 53.

To specify the global Quality Control settings

The settings on the Quality Control tab are the minimum required coverage values (coverage threshold values) that a panel region must meet, or it is flagged as failed region for a run. The coverage values that you specify here are *global settings*, which means that, by default, they are automatically applied to all panel regions in all runs.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the Quality Control tab.

Figure 2-3: Settings dialog box, Quality Control tab



3. Specify the value for the Quality Control settings.
   - Minimum Coverage—The minimum number of reads that are required for a panel region.
   - Average Coverage—The average number of reads that are required for a panel region.
   - Percent Covered (%)—The minimum percentage of the panel region that has reads aligned to it.

4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - "To configure the reference directory" on page 46.
   - "To specify the highlighting colors for variants of a pathogenicity" on page 48.
   - "To apply shared preferences or to clear all tab preferences" on page 51.
   - "To specify variant preferences" on page 53.
   - "To specify Alamut settings" on page 54.
   - "To specify HGMD settings" on page 56.
   - "To specify the Geneticist Assistant password requirements" on page 57.
   - "To specify the OMIM settings" on page 58.
   - "To modify the SoftGenetics license server settings" on page 59.

To specify the highlighting colors for variants of a pathogenicity

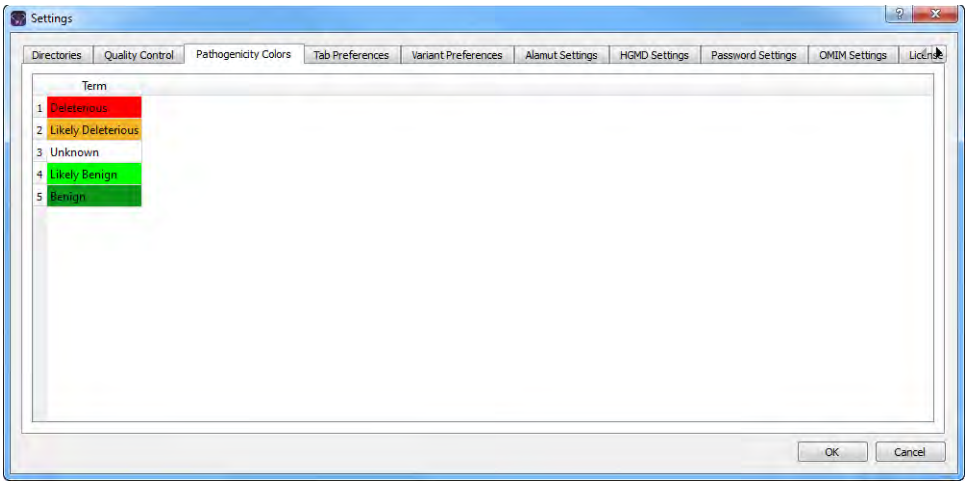
By default, variants for any of the five system-supplied pathogenicity values (Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign) are highlighted in a specific color on the variant sub-tabs and panes. You can use these default colors, or you can specify other colors on the Pathogenicity Colors tab that better meet your business needs.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the Pathogenicity Colors tab.

   The default highlighting colors for the five system-supplied pathogenicity values are displayed on the tab. See Figure 2-4 on page 49.

If you have added a custom pathogenicity value (see "Managing Objects" on page 41), then this value is also displayed, and if applicable, in its selected highlighting color.

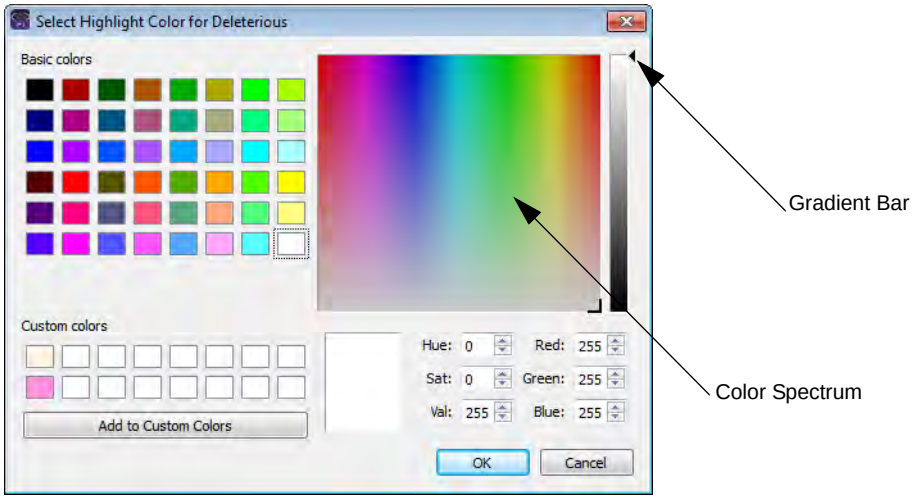
Figure 2-4: Settings dialog box, Default colors shown on the Pathogenicity Colors tab



3. Select a pathogenicity.

   The Select Highlight Color dialog box opens. The pathogenicity that you selected is displayed in the title bar of the dialog box.

Figure 2-5: Select Highlight Color dialog box



4. Continue to one of the following:

   - "To specify a basic highlighting color for a pathogenicity" on page 50.
   - "To specify a custom highlighting color for a pathogenicity" on page 50.

Multiple ways are available for selecting a highlighting color for a pathogenicity. The following are for example purposes only. You can select whichever method or combination of methods best suits your working needs.

To specify a basic highlighting color for a pathogenicity

1. Click a basic color.
2. Click OK.

   The Select Highlight Color dialog box closes. The Pathogenicity Colors tab on the Settings dialog box remains open. The selected pathogenicity is highlighted in the basic color.
3. Continue selecting pathogenicities and specifying basic colors as needed.
4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To apply shared preferences or to clear all tab preferences” on page 51.
   - “To specify variant preferences” on page 53.
   - “To specify Alamut settings” on page 54.
   - “To specify HGMD settings” on page 56.
   - “To specify the Geneticist Assistant password requirements” on page 57.
   - “To specify the OMIM settings” on page 58.
   - “To modify the SoftGenetics license server settings” on page 59.

To specify a custom highlighting color for a pathogenicity

1. Click in a blank custom color box.
2. Click the color in the Color Spectrum to select an approximate color.

   A set of crosshairs is displayed where you clicked and the corresponding color is displayed below the spectrum. The HSV and RGB values are also displayed for the color.
3. Do one of the following:
   - Manually set the HSV or RGB values for the color.
   - Drag and drop the crosshairs to a new location in the Color Spectrum.
   - Point and hold your cursor on the Gradient arrow to the right of the Color Spectrum and move the arrow up and down until the appropriate color is displayed.
4. Click Add to Custom Colors.

5. Click OK.

   The Select Highlight Color dialog box closes. The Pathogenicity Colors tab on the Settings dialog box remains open. The selected pathogenicity is highlighted in the custom color.

6. Continue selecting pathogenicities and specifying custom colors as needed.

7. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:

   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To apply shared preferences or to clear all tab preferences” on page 51.
   - “To specify variant preferences” on page 53.
   - “To specify Alamut settings” on page 54.
   - “To specify HGMD settings” on page 56.
   - “To specify the Geneticist Assistant password requirements” on page 57.
   - “To specify the OMIM settings” on page 58.
   - “To modify the SoftGenetics license server settings” on page 59.

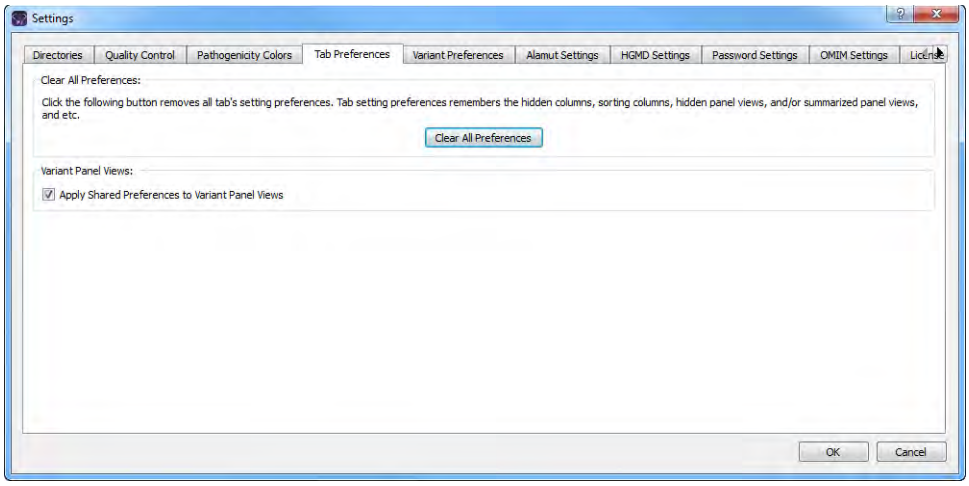
To apply shared preferences or to clear all tab preferences

Remember, a variety of options are available for settings preferences for a tab display, including showing and hiding tab panes, resizing data columns, and so on. (See “Setting Display Preferences in the Geneticist Assistant Main Window” on page 26.) Any preferences that you set for a tab display remain in effect regardless if you log out of and then log back into Geneticist Assistant. You can use an option on the Tab Preferences tab to apply the same column display preferences for variants in all the variant sub-tabs and panes (with the exception of the Sample Group columns on the Sample Variant Data sub-tab on the Sample tab and the Variant pane on the Variant tab) in a single step. You can also use an option on the Tab Preferences tab to clear all the preferred settings for all the data tabs in a single step.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.

2. Open the Tab Preferences tab. See Figure 2-6 on page 52.

Figure 2-6: Settings dialog box, Tab Preferences tab



3. Do one of the following:

   - After you set the column display preferences, select Apply Shared Preferences to Variant Panel Views.

     The same preferences are applied to the appropriate sub-tab or pane on the other tab.

   - To return the tab displays to their default displays (all tab panes shown, all data columns shown, and so on) in a single step, click Clear All Preferences.

     Any customizations that you have made to any and all tabs (hiding tab panes, hiding data columns on tabs, resizing data columns, and so on) are cleared and all tab displays are reset to their default values.

4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:

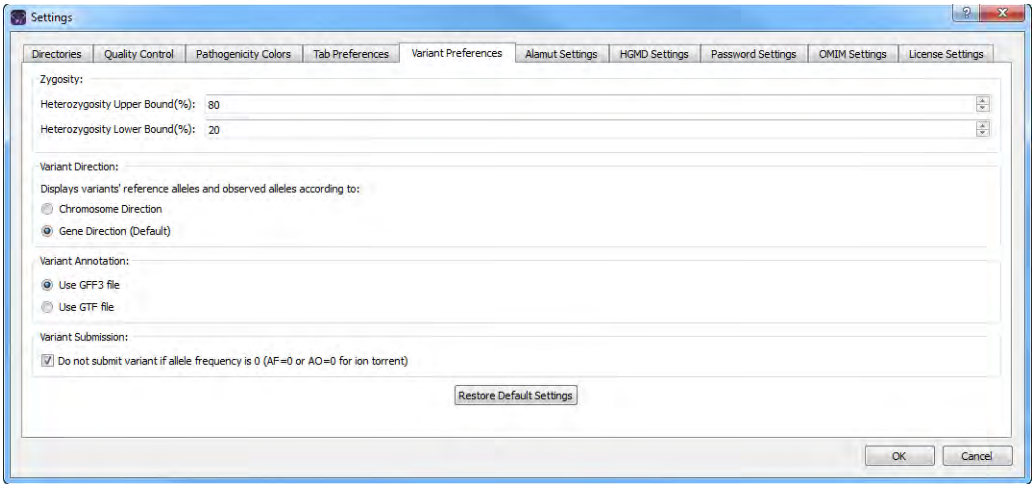
   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To specify the highlighting colors for variants of a pathogenicity” on page 48.
   - “To specify variant preferences” on page 53.
   - “To specify Alamut settings” on page 54.
   - “To specify HGMD settings” on page 56.
   - “To specify the Geneticist Assistant password requirements” on page 57.
   - “To specify the OMIM settings” on page 58.
   - “To modify the SoftGenetics license server settings” on page 59.

To specify variant preferences

Variant preferences determine how the information about variants is displayed on all the variant tabs, sub-tabs, and panes in Geneticist Assistant.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the Variant Preferences tab.

Figure 2-7: Settings dialog box, Variant Preferences tab



3. Set the variant preferences.

Option	Description
Zygosity	
Heterozygosity Upper Bound (%)	Default value is 80. The maximum variant frequency for a variant to be considered heterozygous.
Heterozygosity Lower Bound (%)	Default value is 20. The minimum variant frequency for a variant to be considered heterozygous. **Note:** To report low frequency variants, reduce this value.
Variant Detection	
• Chromosome Direction	• Display the variants' reference alleles and observed alleles based on the positive strand.
• Gene Direction	• Display the variants' reference alleles and observed alleles based on the gene orientation. For a gene on the reverse strand, a reverse complement is used.
Note: To return the Zygosity values and/or Variant Direction value to their default values at any time, click Restore Default Settings.	

Option	Description
Variant Annotation	
• Use GFF3 file	• The default value. Use the annotation information in the reference files that SoftGenetics provides.
• Use GTF file	• Use the annotation information in the reference files that ENSEMBL provides. **Note:** If you select this option, then you must download the correct references files from ENSEMBL and place them in the Reference directory that is defined for your Geneticist Assistant installation. See "To configure the reference directory" on page 46.
Variant Submission	
Do not submit variant if allele frequency is 0	When selected, any variants that are included in a VCF file that have an allele frequency value = 0 in the file are not submitted to the Geneticist Assistant database.

4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - "To configure the reference directory" on page 46.
   - "To specify the global Quality Control settings" on page 47.
   - "To specify the highlighting colors for variants of a pathogenicity" on page 48.
   - "To apply shared preferences or to clear all tab preferences" on page 51.
   - "To specify Alamut settings" on page 54.
   - "To specify HGMD settings" on page 56.
   - "To specify the Geneticist Assistant password requirements" on page 57.
   - "To specify the OMIM settings" on page 58.
   - "To modify the SoftGenetics license server settings" on page 59.

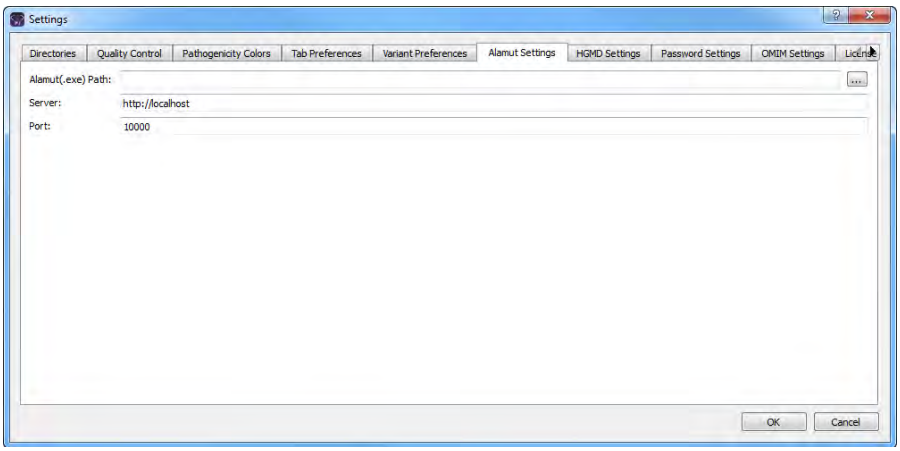
To specify Alamut settings

To view variants that were detected in Geneticist Assistant in Alamut, Geneticist Assistant must be able to connect to Alamut and *Alamut must be installed on the same computer as the Geneticist Assistant client.* You specify the Alamut connection settings on the Alamut Settings tab.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.

2. Open the Alamut Settings tab.

Figure 2-8: Settings dialog box, Alamut Settings tab



3. Click the Browse button ... to open the Select Executable dialog box, and then browse to and select the Alamut executable.

Remember, Alamut must be installed on the same computer as the Geneticist Assistant client.

4. Enter the information for the Alamut server. (The default value is http://localhost).
5. Leave the Port value set to 10000.

Contact SoftGenetics (tech_support@softgenetics.com) for more information about this value, or to change the value.

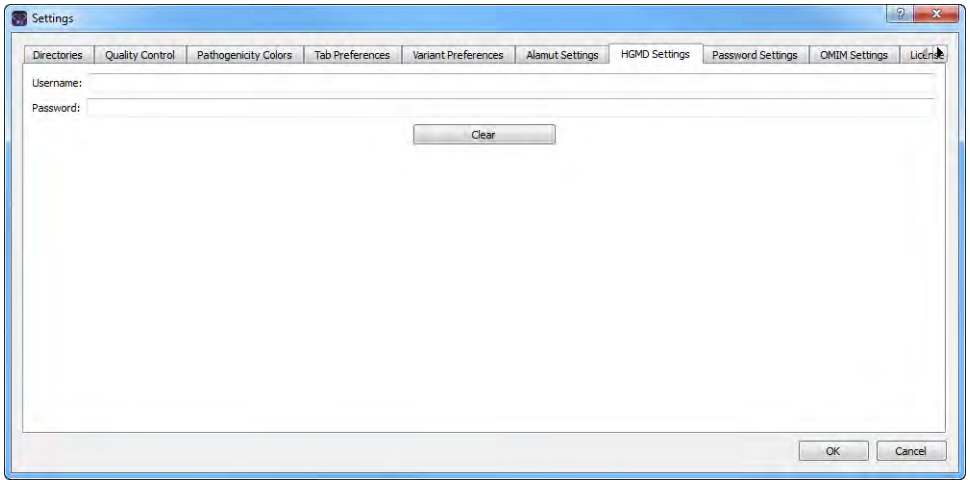
6. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - "To configure the reference directory" on page 46.
   - "To specify the global Quality Control settings" on page 47.
   - "To specify the highlighting colors for variants of a pathogenicity" on page 48.
   - "To apply shared preferences or to clear all tab preferences" on page 51.
   - "To specify variant preferences" on page 53.
   - "To specify HGMD settings" on page 56.
   - "To specify the Geneticist Assistant password requirements" on page 57.
   - "To specify the OMIM settings" on page 58.
   - "To modify the SoftGenetics license server settings" on page 59.

To specify HGMD settings

To view variants that were detected in Geneticist Assistant in HGMD, Geneticist Assistant must be able to connect to HGMD and log you in automatically. You specify your HGMD login settings on the HGMD Settings tab.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the HGMD Settings tab.

Figure 2-9: Settings dialog box, HGMD Settings tab



3. Enter your HGMD username and password.
4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To specify the highlighting colors for variants of a pathogenicity” on page 48.
   - “To apply shared preferences or to clear all tab preferences” on page 51.
   - “To specify variant preferences” on page 53.
   - “To specify Alamut settings” on page 54.
   - “To specify the Geneticist Assistant password requirements” on page 57.
   - “To specify the OMIM settings” on page 58.
   - “To modify the SoftGenetics license server settings” on page 59.

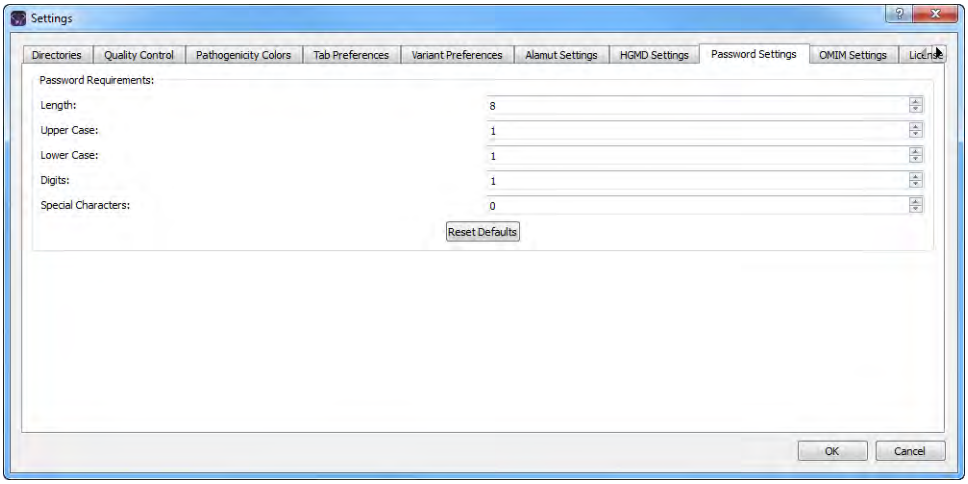
To specify the Geneticist Assistant password requirements

When Geneticist Assistant is first installed, by default, the Username is set to Administrator, and you must specify an administrator password. By default, the only requirement is that the password cannot contain any spaces. You can, however, specify additional requirements that are applicable for all user passwords, including the Administrator password. You can specify the following requirements for your user passwords:

- Minimum length.
- Minimum number of upper case characters.
- Minimum number of lower case characters.
- Minimum number of numeric characters.
- Minimum number of special characters.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the Password Settings tab.

Figure 2-10: Settings dialog box, Password Settings tab



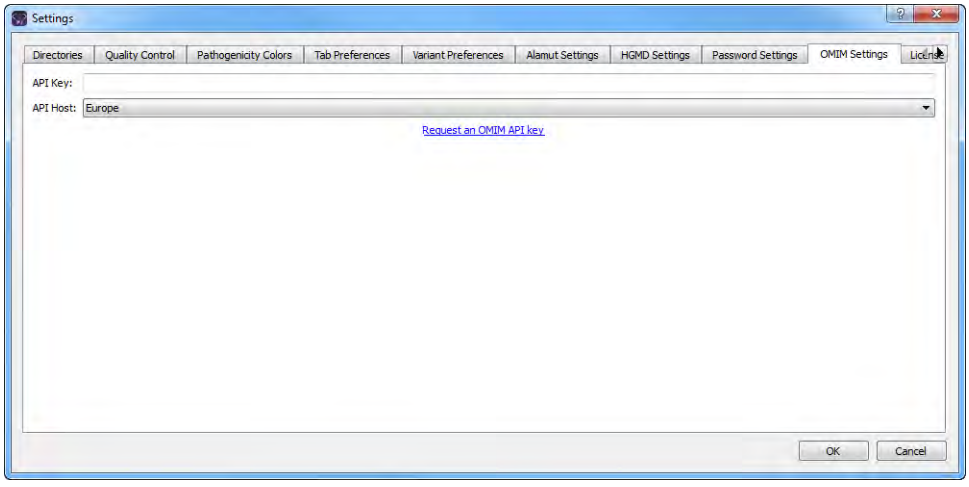
3. Specify the requirements for all user passwords for your Geneticist Assistant installation.
4. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To specify the highlighting colors for variants of a pathogenicity” on page 48.
   - “To apply shared preferences or to clear all tab preferences” on page 51.

To specify the OMIM settings

To view variants that were detected in Geneticist Assistant in OMIM, Geneticist Assistant must be able to connect to OMIM and use an OMIM API key to log you in automatically. You request an OMIM API key and specify the OMIM settings for your Geneticist Assistant installation on the OMIM tab of the Settings dialog box.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the OMIM tab.

Figure 2-11: Settings dialog box, OMIM tab



3. Click Request an OMIM API key.

   You are directed to the API Access to OMIM webpage.
4. Complete the necessary information to obtain an OMIM API Access key.
5. Upon receipt of the key, enter the value in the API field.
6. Indicate the connectivity for the API Host (Europe or United States).

7. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:

   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To specify the highlighting colors for variants of a pathogenicity” on page 48.
   - “To apply shared preferences or to clear all tab preferences” on page 51.
   - “To specify variant preferences” on page 53.
   - “To specify Alamut settings” on page 54.
   - “To specify the Geneticist Assistant password requirements” on page 57.
   - “To modify the SoftGenetics license server settings” on page 59.

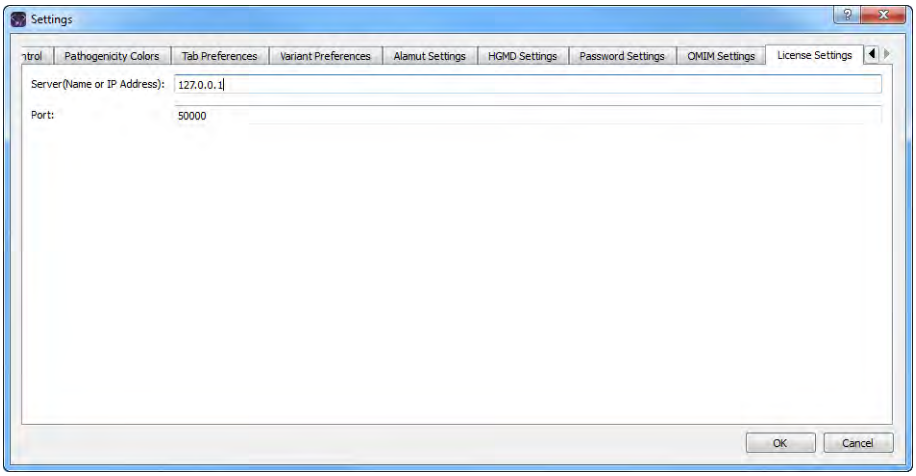
To modify the SoftGenetics license server settings

If you are running a purchased version of Geneticist Assistant, the *first* time that you run the program, you must configure Geneticist Assistant to connect to the SoftGenetics license server. (See “To run a purchased version of Geneticist Assistant” on page 20.) If you change the location of this license server, then you must modify the license server settings so that Geneticist Assistant can still connect to the server.

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the License Settings tab.

   The current information (name or IP address) for the SoftGenetics license server is displayed in the Server field.

Figure 2-12: Settings dialog box, License Settings tab



3. In the server name, enter the name for the server, or its IP address.

If the License Server is running on the same computer on which Geneticist Assistant is running, then you must enter 127.0.0.1.

4. Leave the Port value set to the default value of 50000.
5. If you are done with specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed. See:
   - “To configure the reference directory” on page 46.
   - “To specify the global Quality Control settings” on page 47.
   - “To specify the highlighting colors for variants of a pathogenicity” on page 48.
   - “To apply shared preferences or to clear all tab preferences” on page 51.
   - “To specify variant preferences” on page 53.
   - “To specify Alamut settings” on page 54.
   - “To specify the Geneticist Assistant password requirements” on page 57.
   - “To specify HGMD settings” on page 56.
   - “To specify the OMIM settings” on page 58.

Backing up the Geneticist Assistant Database

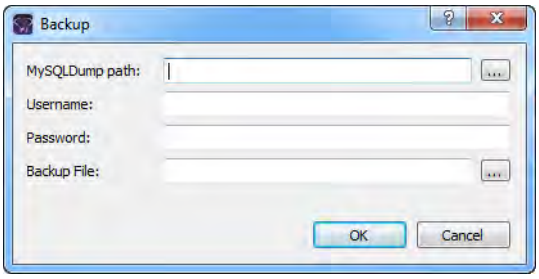
For data protection, you can manually back up the Geneticist Assistant database at any time of your choosing.

To back up the Geneticist Assistant database

1. On the Geneticist Assistant main menu, click Backup > Run Backup.

   The Backup dialog box opens.

Figure 2-13: Backup dialog box



2. Click the Browse button ... next to the MySQLDump path field to open the Select mysqldump.exe dialog box, and then browse to and select the mysqldump.exe file.

If you installed MySQL in is default directory, then the mysqldump.exe file is found in C:\Program Files (x86)\MySQL\MySQL Server 5.1\bin.

3. Enter your Geneticist Assistant Username and password.
4. Click the Browse button ... next to the Backup File field to open the Select backup file dialog box, and specify a name and location for the database backup file.

The default location is C:\Program Files\Softgenetics\Geneticist Assistant\ga_exe, but you can select a different location. The file type is Sql files (.sql) and you cannot change this.*

5. Click OK.

   After the Geneticist Assistant database is successfully backed up, a message opens indicating that the backup is finished.

6. Click OK to close the message.

   The Geneticist Assistant main window remains open.

Chapter 2
Geneticist Assistant Setup and Maintenance

Chapter 3
Geneticist Assistant User Management

In addition to specifying some of the basic settings for your Geneticist Assistant application, such as the Reference directories, or the global Quality Control settings, you must configure the users, and optionally, the groups, who are to have access to the application.

This chapter covers the following topics:

Chapter 3
Geneticist Assistant User Management

Managing Users

Users are the people who log into Geneticist Assistant, whether they are adding and reviewing content, or just using the application in a read-only capacity. Remember, when Geneticist Assistant is first installed, it is installed with a default Administrator user. As the Geneticist Assistant administrator, you must manage all the necessary users for your Geneticist Assistant application, which includes adding new users, deleting users, and modifying user permissions.

Instead of assigning permissions to each user on an individual basis, optionally, you can assign users to a group and apply the same permissions to all the users in the group in a single step. See "Managing Groups" on page 71.

To add a new user

A user in Geneticist Assistant is one of three types:

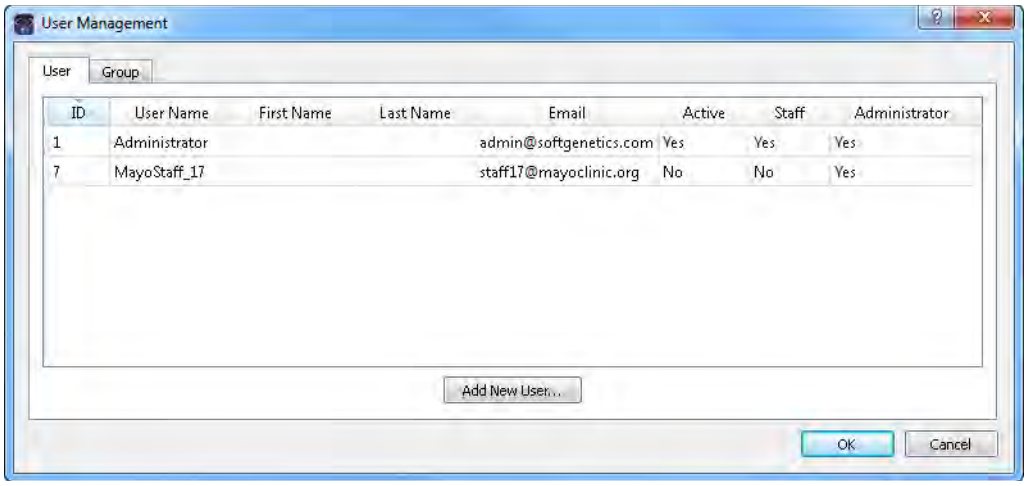
- Administrator user - All available permission types (User, Chemistry, Instrument, and so on) are automatically assigned to an Administrator user. An Administrator user can view all the runs that have been submitted to the Geneticist Assistant database by any user and all the patients that have been added to the Geneticist Assistant database by any user.
- Staff user - A Staff user can log into the django web administrator site. The actions that a staff user can carry out on this site are determined by the permission types that are assigned to the user.
- Standard user - For a "standard" user, you do not select either the Administrator Privileges option or the Staff option. Standard users can view all the runs that they have submitted to the Geneticist Assistant database as well as any runs that have been submitted by other users and for which they have been granted at least the Can Read permission. Standard users can also view all the patients that they have added to the Geneticist Assistantant database as well as any patients that have been added by others users and for which they have been granted at least the Can Read permission.

For a detailed description of the permission types that you can assign to a user, contact SoftGenetics.

1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. By default, the User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant application, their status, and if applicable, their permissions level (Staff or Administrator). See Figure 3-1 on page 66.

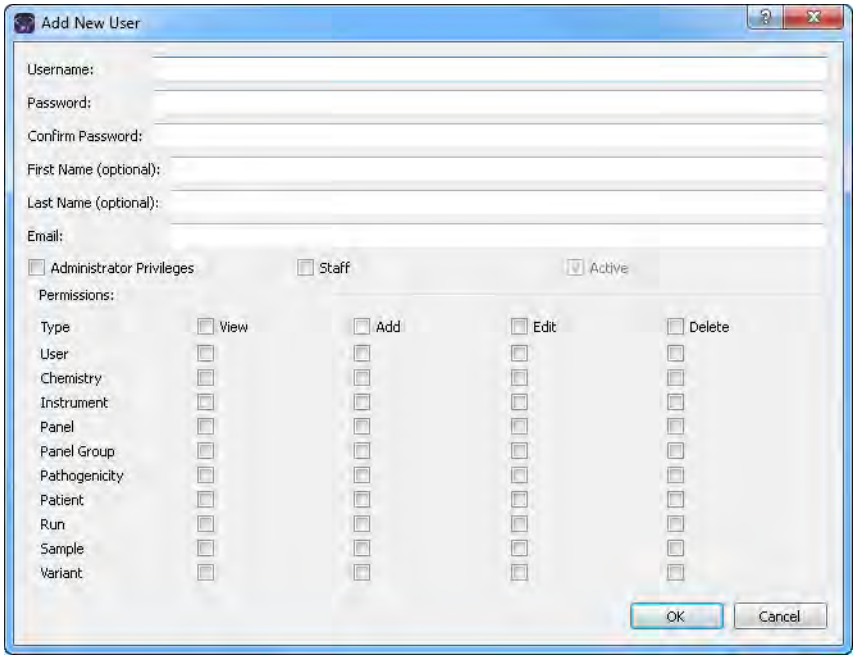
Figure 3-1: User Management dialog box, User tab



2. Click Add New User.

   The Add New User dialog box opens.

Figure 3-2: Add New User dialog box



3. Add the information for the new user.

Field	Description
Username	Required. The login name for the user. Uppercase and lowercase are allowed. No special characters are allowed.
Password	Required. The login password for the user. The password cannot contain any spaces. If additional password requirements have been set, then the password must also adhere to these requirements. See "To specify the Geneticist Assistant password requirements" on page 57.
Confirm Password	Required. Re-enter the password exactly as you entered it in the Password field.
First Name (optional)	The first name for the user. Displayed only here in the application.
Last Name (optional)	The last name for the user. Displayed only here in the application.
Email	Required. The email address for the user.
• Administrator Privileges • Staff	You can assign both Administrator Privileges and Staff status to the same user. Do not assign either option for a standard user.
Active	Selected by default and you cannot change this. The user is added as an Active user. The user can log into Geneticist Assistant and carry out all the activities available to him/her based on the assigned permissions. You can inactivate a user when you edit a user. See "To edit a user" on page 69.

4. In the Permissions pane, add or delete the permissions for the user as needed. You can add or delete permissions individually, or you can add or delete all permissions of a specific type in a single step.
   - To add all permissions of a specific type (for example, View), select the check box next to the type.
   - To clear all permissions of a specific type, clear the check box for the type.

Be aware of the dependencies for these permissions. For example, if a user is to be able to add chemistries, then the user must also be able to view chemistries.

5. Click OK.

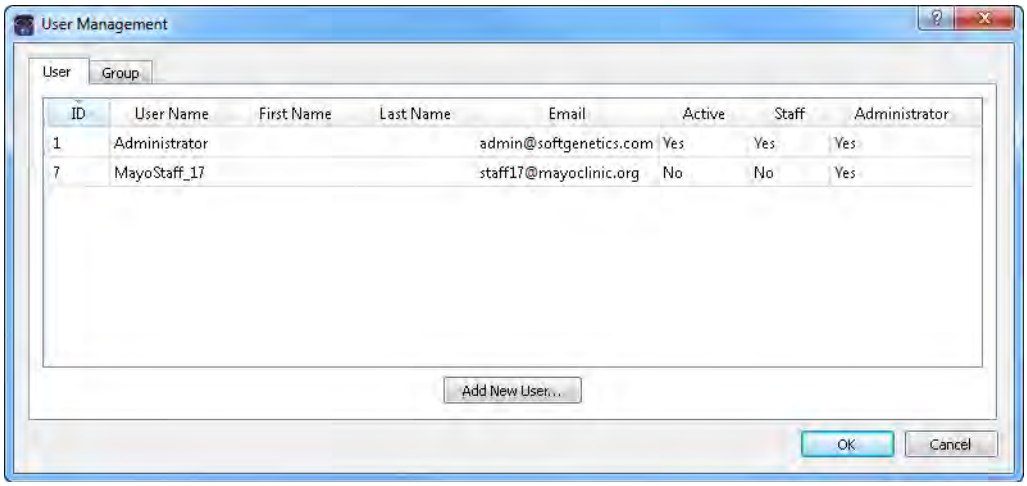
   The User Management dialog box closes. You return to the Geneticist Assistant main window.

To delete a user

1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. By default, the User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant application, their status, and if applicable, their permissions level (Staff or Administrator).

Figure 3-3: User Management dialog box, User tab



No confirmation message opens asking you if you are sure that you want to delete a selected user. Instead, the user is deleted immediately, so be very sure that you want to delete a user before doing so.

2. Right-click the user that you are deleting, and on the context menu that opens, click Remove User.

   The user is deleted.

3. Click OK.

   The User Management dialog box closes. You return to the Geneticist Assistant main window.

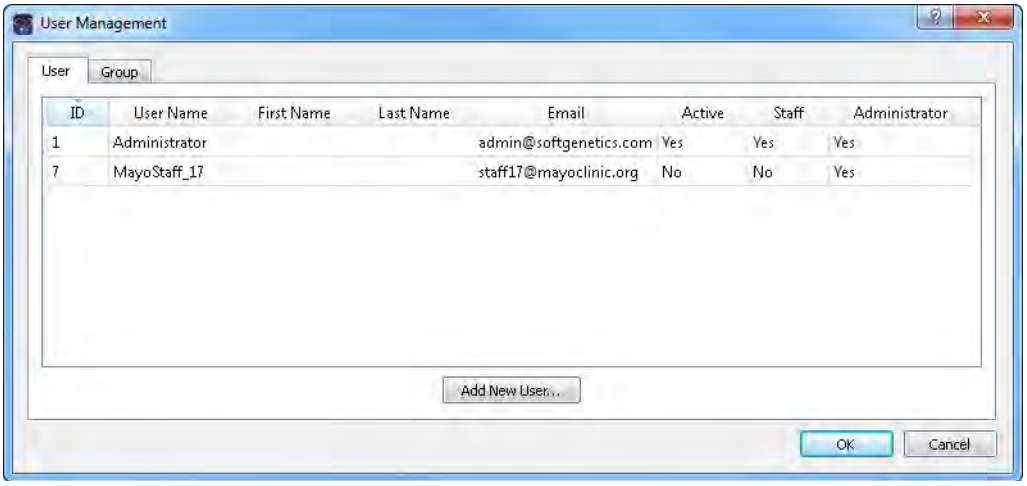
To edit a user

When you edit a user, you can modify the user password, the user first and last names, the user email address, the user status, the user level (Administrator and/or Staff) and the user permissions. You cannot edit a user name. To modify a user name, you must delete the existing user (see "To delete a user" on page 68), and then add the user with a new user name.

1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. By default, the User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant application, their status, and if applicable, their permissions level (Staff or Administrator).

Figure 3-4: User Management dialog box, User tab

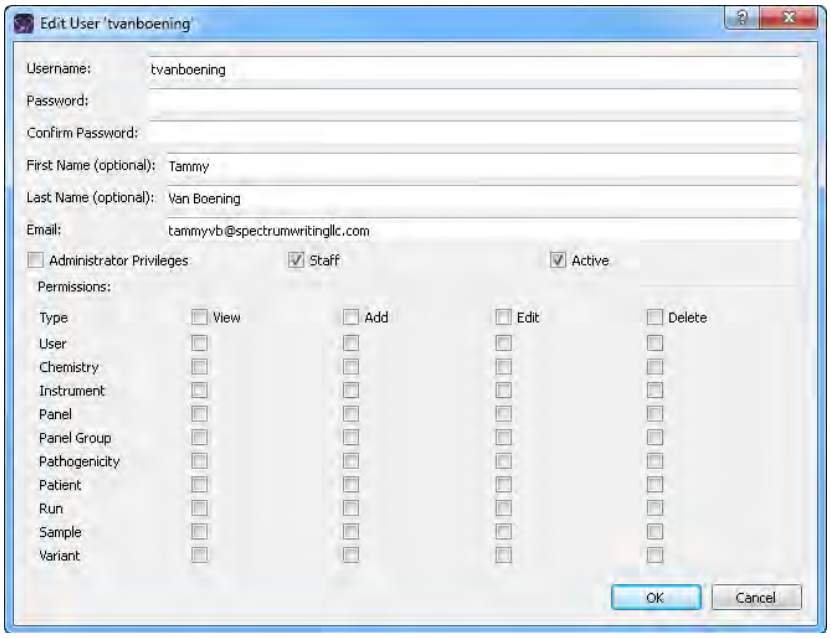


2. Do one of the following:

   - Double-click the user that you are deleting.
   - Right-click the user that you are editing, and on the context menu that opens, click Edit User.

   The Edit User dialog box opens. The dialog displays all the current information for the user, including the currently assigned permissions. See Figure 3-5 on page 70.

Figure 3-5: Edit User dialog box



3. Edit any or all of the following information as needed—the user password, the user first and last names, the user email address, the user status (Active), or the user level (Staff or Administrator).

If you modify the user level, do not forget to manually edit the permissions as needed. See Step 4 below.

4. In the Permissions pane, add or delete the permissions for the user as needed. You can add or delete permissions individually, or you can add or delete all permissions of a specific type in a single step.
   - To add all permissions of a specific type (for example, View), select the check box next to the type.
   - To clear all permissions of a specific type, clear the check box for the type.

Be aware of the dependencies for these permissions. For example, if a user is to be able to add chemistries, then the user must also be able to view chemistries.

5. Click OK.

   The Edit Permissions dialog box closes. You return to the User tab of the User Management dialog box.

6. Click OK.

   The User Management dialog box closes. You return to the Geneticist Assistant main window.

Managing Groups

Instead of assigning permissions to each user on an individual basis, optionally, you can assign users to a *group* and apply the same permissions to all the users in the group in a single step. As the Geneticist Assistant administrator, you must manage all the necessary groups for your Geneticist Assistant application, which includes adding new groups, deleting users, and modifying group permissions.

To add a new group

When you add a group, you must name the group, add to users to the group, and assign permissions to the group. If a user has different privileges assigned than the group permissions, then the complete set of user permissions is the user permissions plus the group permissions. If a user is assigned to multiple groups, then the user's permissions are a sum of all the groups' permissions.

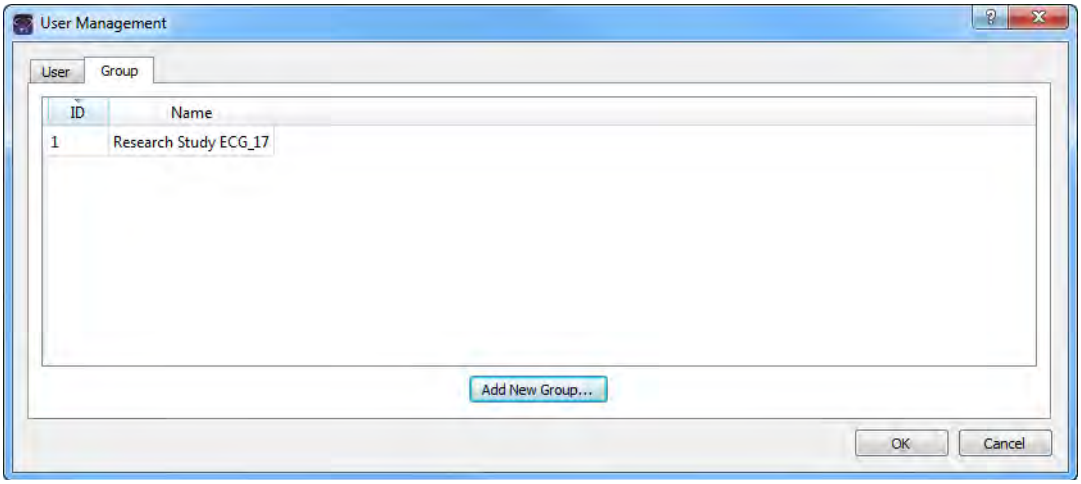
1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. By default, the User tab is the open tab.

2. Click the Group tab to open it.

   The tab displays all the groups by name that are currently configured for your Geneticist Assistant application.

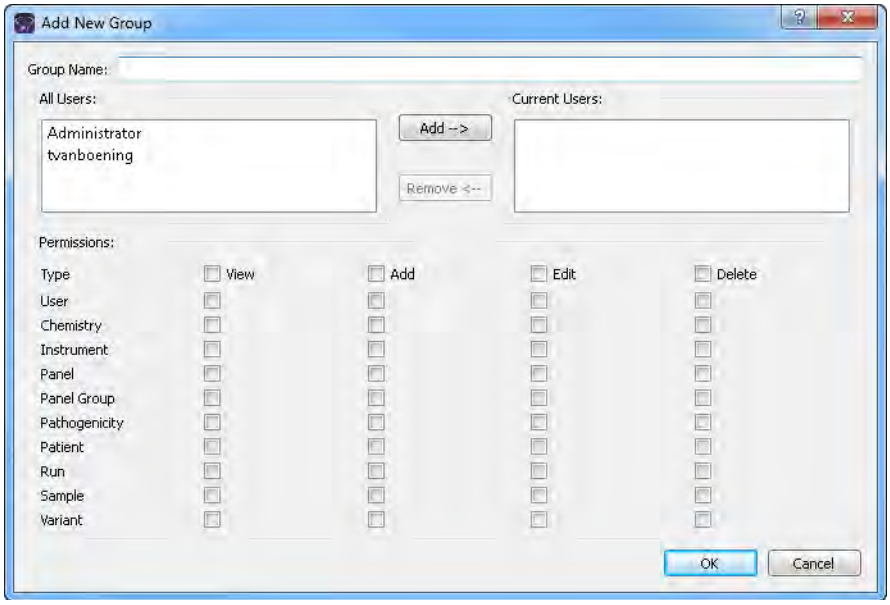
Figure 3-6: User Management dialog box, Group tab



3. Click Add New Group.

   The Add New Group dialog box opens. All users who can be added to a group are displayed in the All Users pane.

Figure 3-7: Add New Group dialog box



4. Enter the name for the new group.

Uppercase and lowercase and spaces are allowed. No special characters are allowed.

5. In the All Users pane, select the users whom you are adding to the group (press and hold the CTRL key to select multiple users in a single step), and then click Add.

If you add a user in error, you can remove them. Select the user in the Current Users pane (press and hold the CTRL key to select multiple users in a single step), and then click Remove.

6. In the Permissions pane, add or delete the permissions for the group as needed. You can add or delete permissions individually, or you can add or delete all permissions of a specific type in a single step.

   - To add all permissions of a specific type (for example, View), select the check box next to the type.
   - To clear all permissions of a specific type, clear the check box for the type.

Be aware of the dependencies for these permissions. For example, if a group is to be able to add chemistries, then the group must also be able to view chemistries.

7. Click OK.

   The Add New Group dialog box closes. The User Management dialog box remains opens.

8. Click OK.

   The User Management dialog box closes. The Geneticist Assistant main window remains opens.

To delete a group

When you delete a group, only the group is deleted. All the users remain configured (with their user permissions, if applicable), in Geneticist Assistant.

No confirmation message opens asking you if you are sure that you want to delete a selected group. Instead, the group is deleted immediately, so be very sure that you want to delete a group before doing so.

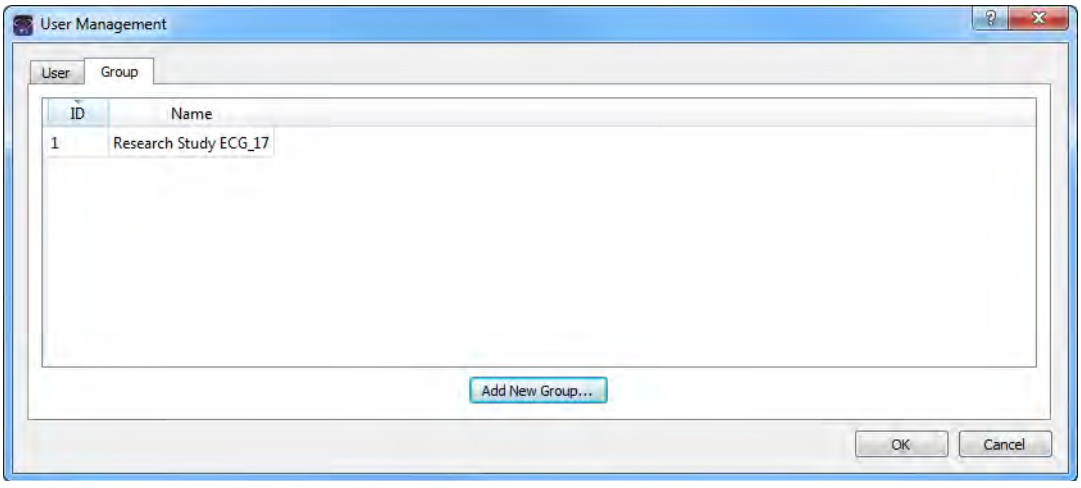
1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. By default, the User tab is the open tab.

2. Click the Group tab to open it.

   The tab displays all the groups by name that are currently configured for your Geneticist Assistant application.

Figure 3-8: User Management dialog box, Group tab



3. Right-click the group that you are deleting, and on context menu that opens, click Remove Group.

   The group is deleted.

4. Click OK.

   The User Management dialog box closes. You return to the Geneticist Assistant main window.

To edit a group

When you edit a group, you can edit the users who belong to the group and the group permissions. You cannot edit the group name. To edit a group name, you must delete the existing group (see "To delete a group" on page 73), and then add the group with a new group name.

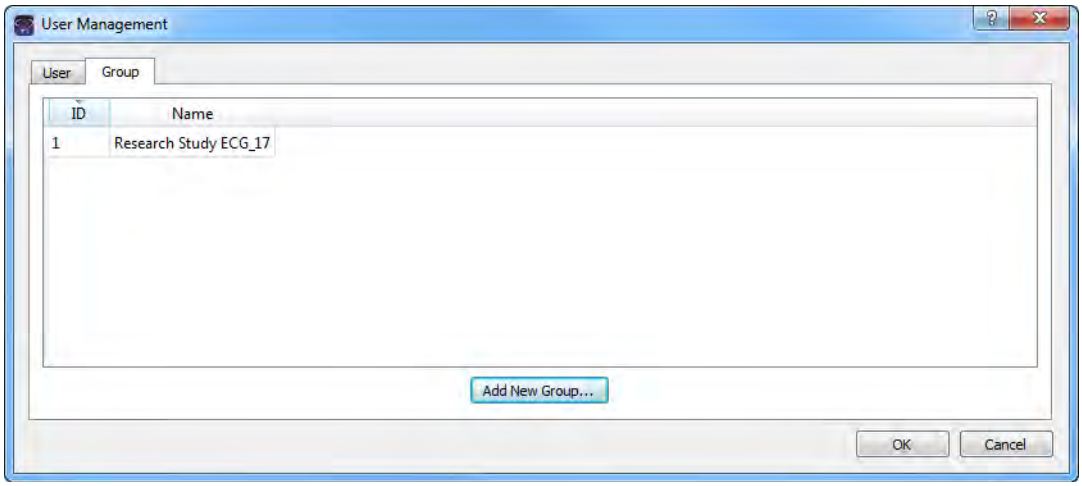
1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. By default, the User tab is the open tab.

2. Click the Group tab to open it.

   The tab displays all the groups by name that are currently configured for your Geneticist Assistant application.

Figure 3-9: User Management dialog box, Group tab

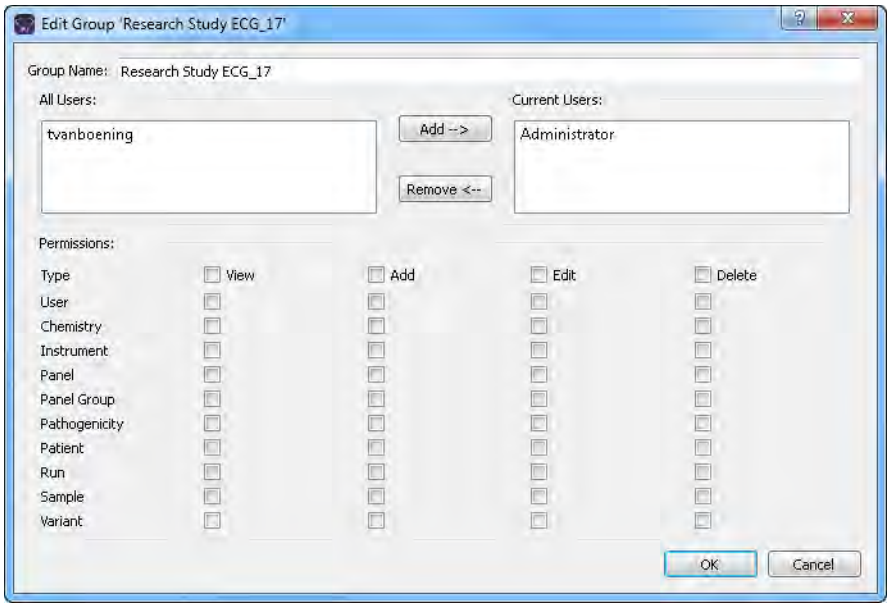


3. Do one of the following:

   - Double-click the group that you are editing.
   - Right-click the group that you are editing, and on the context menu that opens, click Edit Group.

   The Edit Group dialog box opens. The dialog displays all the current information for the group—the group name, the users who are currently assigned to the group, and the permissions for the group. See Figure 3-10 on page 75.

Figure 3-10: Edit Group dialog box



4. To edit the users who belong to the group, do one or both of the following as needed:
   - To add users to the group—In the All Users pane, select the users whom you are adding to the group (press and hold the CTRL key to select multiple users in a single step), and then click Add.
   - To remove users from a group—In the Current Users pane, select the users whom you are removing from the group, (press and hold the CTRL key to select multiple users in a single step), and then click Remove.
5. In the Permissions pane, add or delete the permissions for the group as needed. You can add or delete permissions individually, or you can add or delete all permissions of a specific type in a single step.
   - To add all permissions of a specific type (for example, View), select the check box next to the type.
   - To clear all permissions of a specific type, clear the check box for the type.

Be aware of the dependencies for these permissions. For example, if a group is to be able to add chemistries, then the group must also be able to view chemistries.

6. Click OK.

   The Edit Group dialog box closes. You return to the Group tab of the User Management dialog box.

7. Click OK.

   The User Management dialog box closes. You return to the Geneticist Assistant main window.

Chapter 4
Managing Panels

A *panel* is a collection of genomic regions that are targeted for a specific genomic test or study. Panels are added to Geneticist Assistant by providing a BED file that defines the targeted regions, known as *panel regions*. Each sample that is submitted in Geneticist Assistant is assigned to a specific panel. Managing panels consists of adding, modifying, and deleting panels, and reviewing panel statistics.

This chapter covers the following topics:

Chapter 4
Managing Panels

Adding a Panel

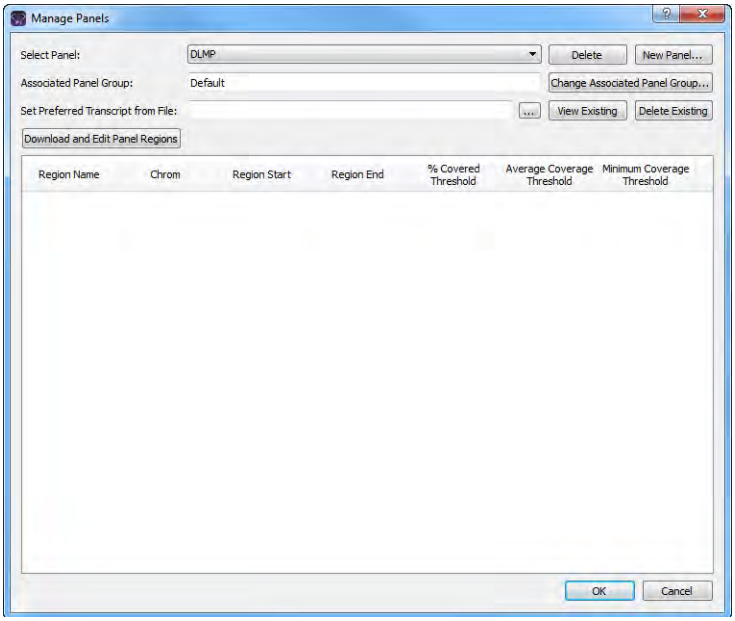
Managing panels includes adding the new panels that will be used for importing your data into Geneticist Assistant.

To add a panel

1. On the Geneticist Assistant main menu, click Panels > Manage Panels.

   The Manage Panels dialog box opens.

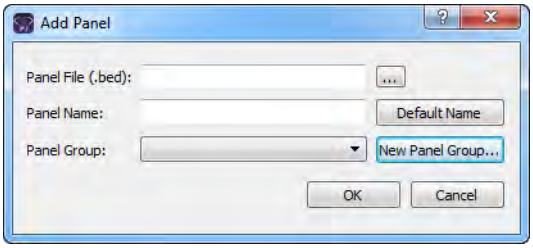
Figure 4-1: Manage Panels dialog box



2. Click New Panel.

   The Add Panel dialog box opens.

Figure 4-2: Add Panel dialog box



3. Click the Browse button ... next to the Panel File (.bed) field, and then browse to and select the BED file for the panel that you are adding.

If the correct BED file is not available for a panel, then you can create a new BED file for it, either from a BED file that was loaded for another panel (see "To create a new BED file by adding or deleting regions" on page 86), or by using the BED File Builder tool to create one based on the information that you specify. See "Building a BED File with the BED File Builder Tool" on page 215.

Geneticist Assistant creates a panel name that is the same as the selected BED file.

4. Optionally, in the Panel Name field, change the name of the panel.

You can always click Default Name to change the name of the panel back to its default name.

5. On the Panel Group dropdown list, select the panel group for the newly added panel.

If the correct panel group is not available, then click New Panel Group to open the Add New Panel Group dialog box and create it. To track variant pathogenicities together for all panels, then instead of creating custom panel groups, create a single panel group to be used for all panels; for example a Global panel group.

6. Click OK.

The Add Panel dialog box closes. The Manage Panels dialog box remains open.

The Manage Panels dialog box display is updated to show all the panel regions by name for the panel. By default, the coverage thresholds that are initially displayed are the global values that are defined on the Quality Control tab on the Settings tab. See "To specify the global Quality Control settings" on page 47.

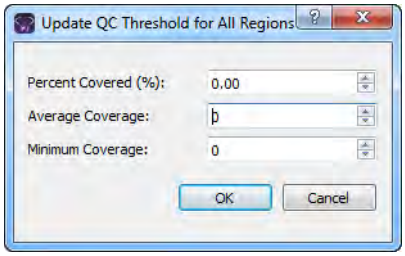
7. If you are done with the configuration of the newly added panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window; otherwise, continue to any of the following as needed:

   - "To manually set new coverage thresholds for panel regions" on page 81.
   - "To create a QC Threshold file for panel regions" on page 82.
   - "To set new coverage thresholds for panel regions with a QC Threshold file" on page 83.
   - "To set or delete a preferred transcript list for a panel" on page 84.
   - "To create a new BED file by adding or deleting regions" on page 86.

To manually set new coverage thresholds for panel regions

After you have created a new panel, the coverage values that are initially displayed for each panel region are the global values that are defined on the Quality Control tab of the Settings dialog box. If needed, you can edit these values and save the values for the panel. You can edit the values for all the regions in a single step, or you can set different coverage thresholds on a per region basis.

1. If you have just created a new panel, then the Manage Panels dialog box is already open and the panel regions are displayed; otherwise, do the following:
   a. On the Geneticist Assistant main menu, click Panels > Manage Panels to open the Manage Panels dialog box.
   b. On the Select Panel dropdown list, select the appropriate panel.
   c. Click Download and Edit Panel Regions to display the panel regions on the Manage Panels dialog box.
2. Do one of the following:

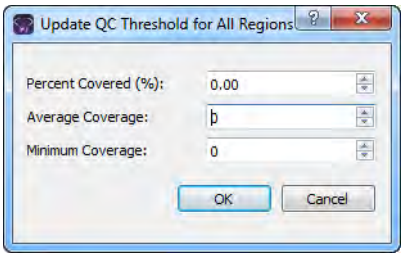
Action	Steps
To edit the values for all regions in a single step	1. Click Update QC Threshold. The Update QC Threshold for All Regions dialog box opens. *Figure 4-3: Update QC Threshold for All Regions dialog box*  2. For each coverage value that is to be edited, double-click it to select it, and then manually enter a new value, or click the Up/Down arrows to change the value accordingly. 3. Click OK.
To edit the values on a per region basis	1. Double-click in the appropriate coverage field for a region. The coverage value is selected, and Up/Down arrows are displayed. 2. Manually enter a new value, or click the Up/Down arrows to change the value accordingly.

3. If you are done with the configuration of the newly added panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. (The edited coverage values are now saved for the panel in the Geneticist Assistant database.) Otherwise, continue any of the following as needed:
   - "To create a QC Threshold file for panel regions" below.
   - "To set or delete a preferred transcript list for a panel" on page 84.
   - "To create a new BED file by adding or deleting regions" on page 86.

To create a QC Threshold file for panel regions

After you edit the coverage values for the regions of one panel, you can save these edited values to a QC Threshold text file. You can save this file as record of the QC thresholds that were used for the panel and you can also load this text file to apply these coverage values to a different panel.

1. If you have just created a new panel, then the Manage Panels dialog box is already open and the panel regions are displayed; otherwise, do the following:
   a. On the Geneticist Assistant main menu, click Panels > Manage Panels to open the Manage Panels dialog box.
   b. On the Select Panel dropdown list, select the appropriate panel.
   c. Click Download and Edit Panel Regions to display the panel regions on the Manage Panels dialog box.
2. Do one of the following:

Action	Steps
To edit the values for all regions in a single step	1. Click Update QC Threshold. The Update QC Threshold for All Regions dialog box opens. *Figure 4-4: Update QC Threshold for All Regions dialog box*  2. For each coverage value that is to be edited, double-click it to select it, and then manually enter a new value, or click the Up/Down arrows to change the value accordingly. 3. Click OK.

Action	Steps
To edit the values on a per region basis	1. Double-click in the appropriate coverage field for a region. The coverage value is selected, and Up/Down arrows are also displayed. 2. Manually enter a new value, or click the Up/Down arrows to change the value accordingly.

3. Click Save QC Threshold File.

   The Save File dialog box opens. By default, the QC Threshold file is named as <Panel Name>_qc_threshold, and it is saved in the same directory as the panel folder. You can change one or both of these values. The file type is a text file, and you cannot change this.

4. Optionally, change the name for the QC Threshold file, and/or the directory in which to save the file, and then click Save.

   The Save File dialog box closes. The Manage Panels dialog box remains open. You can now load this text file to apply these coverage values to a different panel. See "To set new coverage thresholds for panel regions with a QC Threshold file" below.

5. If you are done with the configuration of the newly added or edited panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. (The edited coverage values are now saved for the panel in the Geneticist Assistant database.) Otherwise, continue to one or both of the following as needed:

   - "To set or delete a preferred transcript list for a panel" on page 84.
   - "To create a new BED file by adding or deleting regions" on page 86.

To set new coverage thresholds for panel regions with a QC Threshold file

After you edit the coverage values for the regions of one panel, you can save these edited values to a QC Threshold text file, and then load this text file to apply these coverage values to a different panel.

1. If you have just created a new panel, then the Manage Panels dialog box is already open and the panel regions are displayed; otherwise, do the following:

   a. On the Geneticist Assistant main menu, click Panels > Manage Panels to open the Manage Panels dialog box.

   b. On the Select Panel dropdown list, select the appropriate panel.

   c. Click Download and Edit Panel Regions to display the panel regions on the Manage Panels dialog box.

2. Click Load QC Threshold from File.

   The Select a file to set QC threshold dialog box opens.

3. Browse to and select the appropriate QC Threshold file, and then click Open.

   The Select a file to set QC threshold dialog box closes, and a QC Threshold Updated message opens. The message indicates the number of regions for which the QC threshold values were successfully updated.

If the correct QC Threshold file is not available, then you can create it. See "To create a QC Threshold file for panel regions" on page 82.

4. Click OK.

   The message closes. The Manage Panels dialog box remains open.

5. If you are done with the configuration of the newly added or edited panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. (The edited coverage values are now saved for the panel in the Geneticist Assistant database.) Otherwise, continue to one or both of the following as needed:

   - "To set or delete a preferred transcript list for a panel" on page 84.
   - "To create a new BED file by adding or deleting regions" on page 86.

To set or delete a preferred transcript list for a panel

A *preferred transcript* list is a tab-delimited text file that defines the specific transcript or isoform that is to be used for a gene. You can use different transcripts for different panels. The preferred transcript list contains two fields. The first column contains the gene name. The second column specifies the transcript by its nm_accession number. There are no restrictions for the file name, but you must save the file as a tab-delimited text file.

1. If you have just created a new panel, then the Manage Panels dialog box is already open and the panel regions are displayed; otherwise, do the following:

   a. On the Geneticist Assistant main menu, click Panels > Manage Panels to open the Manage Panels dialog box.

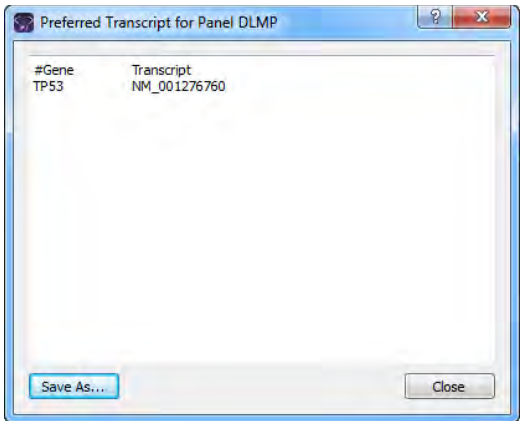
   b. On the Select Panel dropdown list, select the appropriate panel.

2. Optionally, do any or all of the following as needed:

   - To view the transcript list that is currently set as the preferred transcript list for the panel, click View Existing.

     The Preferred Transcript for <Panel Name> dialog box opens. The dialog box displays the transcript list that is currently set as the preferred list for the panel. See Figure 4-5 on page 85.

Figure 4-5: Preferred Transcript dialog box



- To save the preferred transcript list with a different name, click Save As in the Preferred Transcript dialog box; otherwise, click Close to close the dialog box.

  The Manage Panels dialog box remain open.

The Save As option provides a quick way of copying the preferred transcript list so that you can make any needed changes to it, and then use it with this panel or another panel.

- To set a preferred transcript list for the panel, click the Browse button ... next to the Set Preferred Transcript from File field, and in the Open File dialog box, browse to and select the appropriate transcript list, and then click Open.

  The Open File dialog box closes. The full path to the selected transcript list is displayed in the Set Preferred Transcript from File field.

- To delete the preferred transcript list for the panel, click Delete Existing, and then answer Yes in the Delete Existing Preference message.

  The Manage Panels dialog box remain open.

3. If you are done with the configuration of the newly added or edited panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. Otherwise, continue to any of the following as needed:

   - “To manually set new coverage thresholds for panel regions” on page 81.
   - “To create a QC Threshold file for panel regions” on page 82.
   - “To set new coverage thresholds for panel regions with a QC Threshold file” on page 83.
   - “To create a new BED file by adding or deleting regions” on page 86.

To create a new BED file by adding or deleting regions

Sometimes, you might need to use a panel that is a combination of two or more BED files. After you load and save a BED file for a panel, you can add regions from an existing BED file to it and then save this modified BED file as a new BED file. You can also delete regions from the loaded BED file and save this modified BED file as a new BED file.

When you add regions from an existing BED file to another BED file, all the regions are added from the existing BED file. You cannot select specific regions to add; however, after all the regions are added, you can delete one or more regions.

1. If you have just created a new panel, then the Manage Panels dialog box is already open and the panel regions are displayed; otherwise, do the following:
   a. On the Geneticist Assistant main menu, click Panels > Manage Panels to open the Manage Panels dialog box.
   b. On the Select Panel dropdown list, select the appropriate panel.
   c. Click Download and Edit Panel Regions to display the panel regions on the Manage Panels dialog box.
2. Do one or both of the following as needed:

Action	Steps
To add regions from an existing BED file	1. Click Browse button ... that is next to the Add Regions from BED File field. The Select a file to add to the panel dialog box opens. 2. Browse to and select the appropriate BED file, and then click Open to close the dialog box and return to the Manage Panels dialog box. 3. Click Add Regions.
To delete regions from the loaded BED file	1. Click the region that is to be deleted. (CTRL-click to select multiple regions.) 2. Right-click the selected regions, and on the context menu that opens, click Remove Selected Regions. **Note:** Deleting regions from a BED file does not delete the panel. To delete the panel itself, see "Deleting a Panel" on page 90.

3. On the Manage Panels dialog box, click Save Panel Regions to a BED file.

   The Save File dialog box opens. By default, the new BED file is named the same as the loaded BED file, and it is saved in the same directory as the loaded BED file. You can change one or both of these values.

4. Optionally, change the name for the new BED file, and/or the directory in which to save the file, and then click Save.

   The Save File dialog box closes. The Manage Panels dialog box remains open.

5. If you are done with the configuration of the newly added or edited panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. Otherwise, continue to any of the following as needed:

   - “To manually set new coverage thresholds for panel regions” on page 81.
   - “To create a QC Threshold file for panel regions” on page 82.
   - “To set new coverage thresholds for panel regions with a QC Threshold file” on page 83.
   - “To set or delete a preferred transcript list for a panel” on page 84.

Modifying a Panel

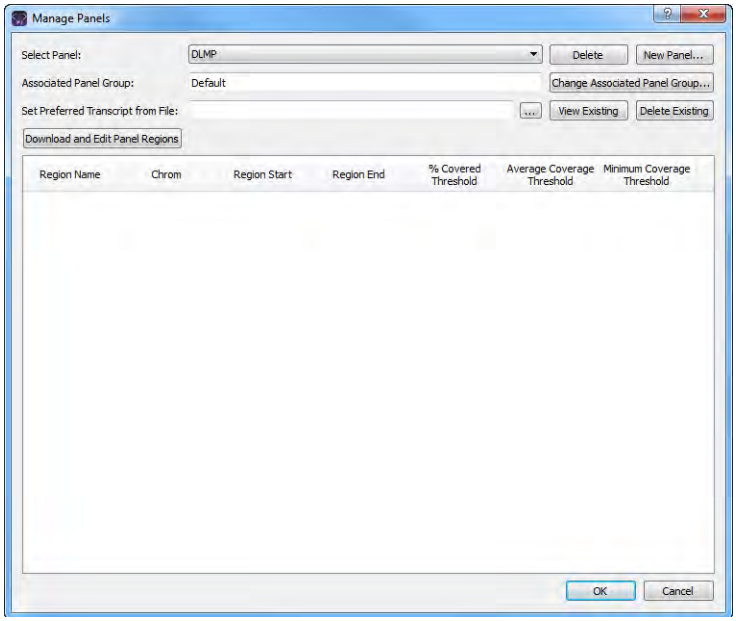
Managing panels includes modifying panels that will be used for importing your data into Geneticist Assistant.

To modify a panel

1. On the Geneticist Assistant main menu, click Panels > Manage Panels.

   The Manage Panels dialog box opens.

Figure 4-6: Manage Panels dialog box



2. On the Select Panel dropdown list, select the panel that is to be modified.
3. Click Download and Edit Panel Regions.

   The panel regions are displayed on the Manage Panels dialog box.
4. Do any or all of the following as needed:
   - To change the associated panel group, click Change Associated Panel Group, and in the Panel Group Association dialog box, make the necessary change.
   - To manually set new coverage thresholds for the panel regions, see “To manually set new coverage thresholds for panel regions” on page 81.
   - To set new coverage thresholds for the panel regions with a QC Threshold file, see “To set new coverage thresholds for panel regions with a QC Threshold file” on page 83.

- To set a preferred transcript list for the panel, or delete a preferred transcript list, see “To set or delete a preferred transcript list for a panel” on page 84.
- To create a new BED file for the panel, see “To create a new BED file by adding or deleting regions” on page 86.

Deleting a Panel

Managing panels includes deleting panels that are no longer needed in Geneticist Assistant.

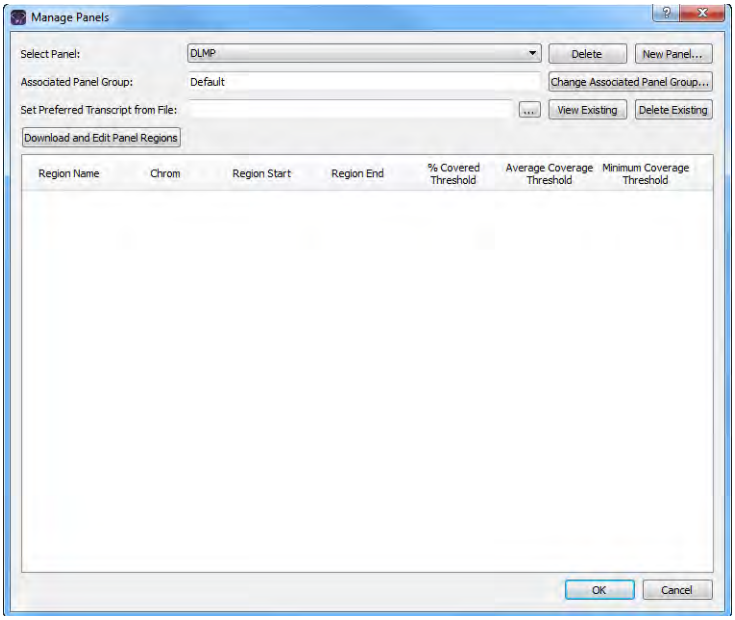
When you delete a panel, you delete the entire panel. To simply delete regions from a BED file, not the entire panel, see *"To create a new BED file by adding or deleting regions" on page 86.*

To delete a panel

1. On the Geneticist Assistant main menu, click Panels > Manage Panels.

   The Manage Panels dialog box opens.

Figure 4-7: Manage Panels dialog box



2. On the Select Panel dropdown list, select the panel that is to be deleted.
3. Click Delete.

   A message opens asking you if you are sure that you want to delete the selected panel.

4. Click Yes.

   The message closes. The entire panel is deleted, and a second message opens indicating that the panel was successfully deleted.

5. Click OK.

   The message closes. The Manage Panels dialog box remains open.

Reviewing Panel Statistics

The Panel Statistics tab has options for reviewing specific information for a selected panel/ panel group and a selected date range. The default date range is for samples that have been added within the last 30 days of the current day's date. You can modify the start and end dates for the date range.

To review panel statistics

1. On the Geneticist Assistant main menu, click Panels > Panel Statistics.

   The Panel Statistics tab opens for the selected panel and panel group.

2. Change any of the display options for the Panel Statistics tab, and then click Refresh to update the display accordingly.

The Panel Statistics tab that opens from the Panels menu is identical to the Panel Statistics tab that opens from the context menu of the Run tab. For a complete description of this tab, see "Panel Statistics tab (Run tab)" on page 164.

Chapter 5
Managing Patients and Runs

A *patient* refers to an individual from whom a sample was taken. A *run* refers to loading a BAM or DAT file *and* a VCF file or just a VCF file into Geneticist Assistant for the purposes of importing a list of variants into the Geneticist Assistant database and/or reviewing a list of variants in a sample. This chapter details how to manage patient settings for your runs and how to add a new run in Geneticist Assistant.

This chapter covers the following topics:

For information about managing your run settings (chemistries, instruments, and so on), see "Managing Objects" on page 41.

Chapter 5
Managing Patients and Runs

Accessing and Reviewing Patient Records

In addition to managing settings (chemistries, instruments, and so on) for your runs, you can manage the patients for your runs. A *patient* refers to an individual from whom a sample was taken. Managing patients for your runs consists of a variety of different activities, including reviewing and modifying information (such as adding comments and adding relationships) for existing patients and related patients, and generating, and optionally, saving, Patient reports to the Geneticist Assistant database.

To review existing patient information

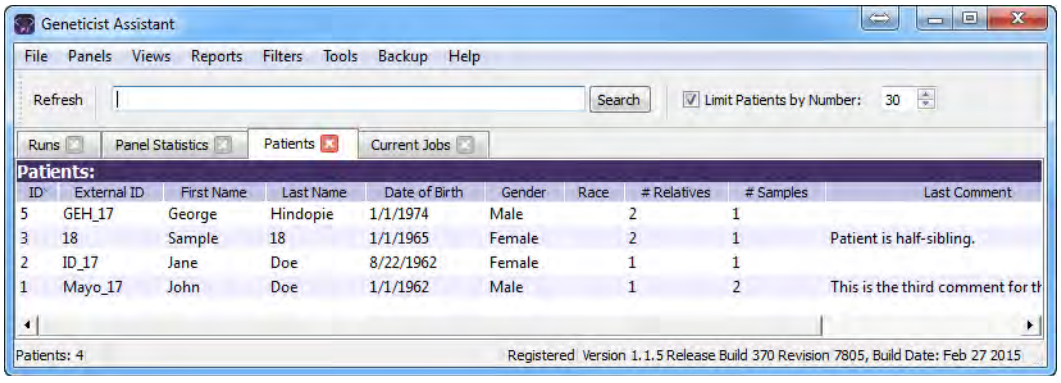
Information for an existing patient is displayed on the Patient tab. When you review the information for an existing patient, a variety of options are available for working with the patient, such as modifying the identifying information for a patient, and adding relationships and comments for the patient.

1. On the Geneticist Assistant main menu, click File > Patients.

   The Patients tab opens.

   - If you are an Administrator user, then the tab can display all the patients that have been added by all users to your Geneticist Assistant database. By default, when the tab first opens, only up to the 30 most recently added patients are displayed and the patients are sorted based on Patient ID.
   - If you are a standard user, then the tab can display all the patients that you have added to the Geneticist Assistant database as well as all the patients that others users have added to the database and for which you have been granted at least the Can Read permission. By default, when the tab first opens, only up to the 30 most recently added patients that fit one of these two criteria are displayed and the patients are sorted based on Patient ID.

Figure 5-1: Patients tab



2. Optionally, do any or all of the following as needed:
   - Change the number of patients by which to limit the display.
   - If you are an Administrator user, clear the Limit Patients by Number option to display all the patients that have been added to your Geneticist Assistant database.
   - If you are a standard user, clear the Limit Patients by Number option to display all the patients you have added to the Geneticist Assistant database as well as all the patients that have been added by other users and for which you have been granted the Read permission.
   - Change the sort order of the patients on the tab. (See "Change the sort order of data" on page 35.)
   - Right-click a patient on the tab to open a context menu with the following options:

Table 5-1: Patient tab context menu

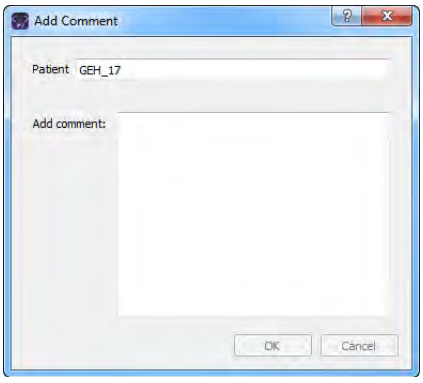
Option	Description
Patient Detail	Opens the Patient tab. See Step 3 below.
Add New Relationships	Opens the Add New Patient dialog box so that you can add a relationship to the selected patient. By default, the Is Related to Existing Patient option is selected and unavailable. Repeat Step 3 of "To add new patients one at a time" on page 118 as many times as needed to add all the necessary patient relationships to the patient.
Remove Patient's Relationships	Removes all the relationships that have been added for the patient.
Delete Patient	Deletes the selected patient from the Geneticist Assistant database.
Add All to Custom Report	Selects all the data that is displayed for the patients for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Add Comment	Opens the Add Patient Comment dialog box. You add a comment for the selected patient in the Add Comment field. The comment is displayed in the Last Comment field and in the Comments pane on the Patient sub-tab on the Patient tab. See "Patient tab, Patient sub-tab, Comments pane" on page 103. *Figure 5-2: Add Patient Comment dialog box*  **Note:** If multiple comments have been added for a patient, then the comment that was added last is displayed in the Last Comment field.

Table 5-1: Patient tab context menu (Continued)

Option	Description
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. If you are an Administrator user, then you can set or modify the permissions for a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See "To set or modify the Read/Write permissions for a patient record" on page 98.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the tab. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

3. Do one of the following to open the Patient tab:

   - Right-click the patient record that you are reviewing, and on the context menu that opens, click Patient Detail.
   - Double-click the patient record that you are reviewing.

   The Patient tab has three sub-tabs. Each sub-tab has different panes for displaying and working with patient information.

Table 5-2: Patient sub-tabs on the Patient tab

Sub-tab	Available Options
Patient sub-tab - See "Patient tab, Patient sub-tab" on page 99.	
	Patient pane • Modifying the identifying information for a patient. • Adding comments for the patient. • Adding relationships to the patient. • Removing relationships for a patient.
	Related Patients pane • Adding relationships to a related patient. • Removing relationships for a related patient.
	Comments pane • Adding comments for a patient. • Deleting comments for a patient.
	Reports pane • Viewing an onscreen preview of a Patient report that was saved to the Geneticist Assistant database. • Deleting a Patient report that was saved to the Geneticist Assistant database.

Table 5-2: Patient sub-tabs on the Patient tab

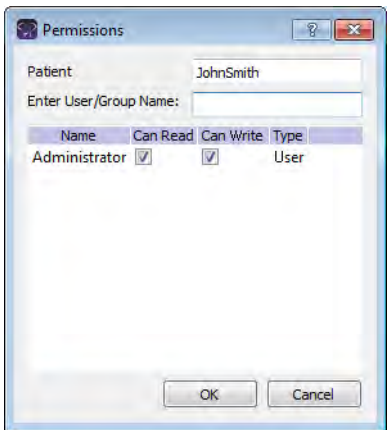
Sub-tab	Available Options
Samples sub-tab - See "Patient tab, Samples sub-tab" on page 106.	
	Samples pane • Viewing the information for the samples that are associated with the patient. • Adding or editing a comment for a sample.
Variants sub-tab - See "Patient tab, Variants sub-tab" on page 111.	
	Variants pane • Viewing a list of all the variants in all the samples that are associated with the patient. • Adding or editing a comment for a variant. • Selecting a different transcript for a variant. • Editing the type field, zygosity field, and/or the HGVS information (Genomic, Coding, and/or Protein) for a variant. • Viewing a variant in Alamut. • Viewing the PubMed abstract for a variant. • Specifying the pathogenicity and pathogenicity status for a variant. • Specifying the artifact type for a variant.

To set or modify the Read/Write permissions for a patient record

1. If you have not already done so, right-click the patient record on the Patients tab, and on the context menu that opens, click Permissions.

   The Permissions dialog box opens. The first time that the dialog box opens, if the patient was added using a Geneticist Assistant version with a build date of 4/8/15 or later, then the name of the user who added the patient is displayed in the lower pane of the dialog box and both the Can Read and Can Write permissions are selected. If a standard user added the patient, then the Administrator user name is also displayed. For patients who were added using earlier versions of Geneticist Assistant, the pane is blank. For subsequent openings, the dialog box displays the names of all the users and/or groups and their assigned permissions for the patient record.

Figure 5-3: Permissions dialog box (Patient tab)



2. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to Step 3.

You can add or remove the Can Write permission for a user or group, or you can remove both the Can Read and Can Write permissions. Removing the Can Read permission but leaving the Can Write permission selected for a user or group has no effect.

3. Enter a user name (the login name for the user), or to assign the same permissions to multiple users in a single step, enter a group name.

   As you enter a user name or a group name, a dropdown list opens. The list is dynamically updated with user names or group names that are in your Geneticist Assistant database and that match the search string that you are entering.

4. Select the appropriate user name or group name from the list, and then press [ENTER].

   The user name or group name is displayed in the lower pane of the dialog box. The Read permission is already assigned for the selected user or group. If just this permission is selected, then the user or group can simply view the patient record. They cannot modify it.

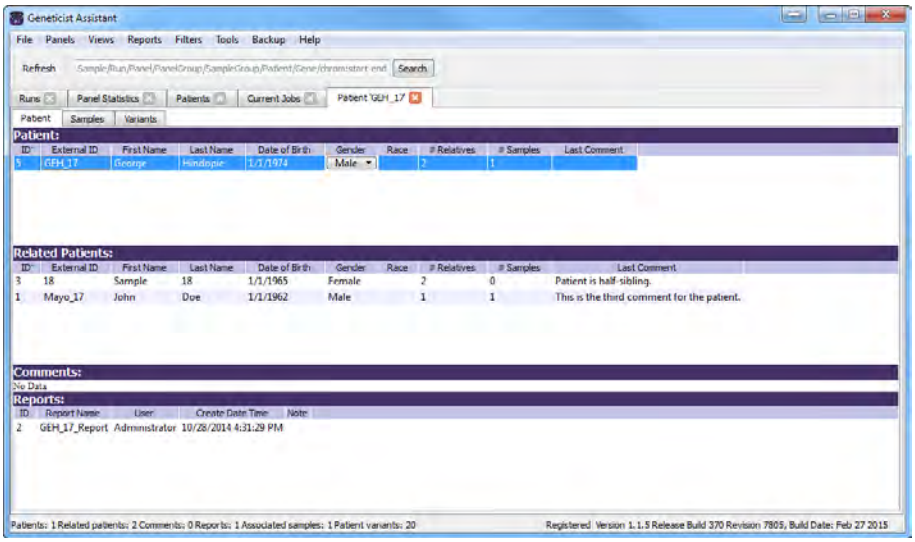
5. Optionally, if the user or group must be able to modify the patient record as well, then select the Can Write permission.

6. Click OK.

Patient tab, Patient sub-tab

When a Patient tab first opens, the Patient sub-tab is the active tab. The tab has four panes that display information about the patient—the Patient pane, the Related Patients pane, the Comments pane, and the Reports pane.

Figure 5-4: Patient tab, Patient sub-tab



See:

- "Patient tab, Patient sub-tab, Patient pane" below.
- "Patient tab, Patient sub-tab, Related Patients pane" on page 102.
- "Patient tab, Patient sub-tab, Comments pane" on page 103.
- "Patient tab, Patient sub-tab, Reports pane" on page 105.

Patient tab, Patient sub-tab, Patient pane

The Patient pane on the Patient sub-tab shows the identifying information for the patient (ID, name, and so on). You can double-click in any of the following fields to modify the information as needed (External ID, First Name, Last Name, and/or Date of Birth) and you can select a different value for the Gender.

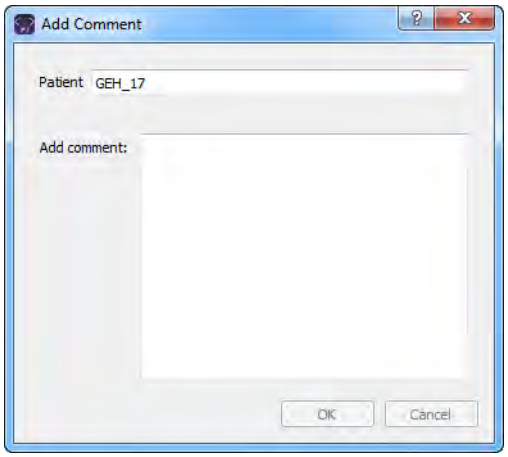
If you are changing the Date of Birth for the patient, then you must select one value (month, day, or year) at a time, and then you can manually enter a a new value, or you can use the Up/Down arrows to change the value.

You can right-click in the pane to open a context menu with the following options:

Table 5-3: Patient tab, Patient sub-tab, Patient pane context menu

Option	Description
Add New Relationships	Opens the Add New Patient dialog box so that you can add a relationship to the selected patient. By default, the Is Related to Existing Patient option is selected and unavailable. Repeat Step 3 of "To add new patients one at a time" on page 118 as many times as needed to add all the necessary patient relationships to the patient.
Remove Patient's Relationships	Removes all the relationships that have been added for the patient.
Add All to Custom Report	Selects all the data that is displayed for the patient for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.

Table 5-3: Patient tab, Patient sub-tab, Patient pane context menu (Continued)

Option	Description
Add Comment	Opens the Add Comment dialog box. You add a comment for the selected patient in the Add Comment field. The comment is displayed in the Last Comment field in the pane and in the Comments pane. See "Patient tab, Patient sub-tab, Comments pane" on page 103. *Figure 5-5: Add Patient Comment dialog box*  **Note:** If multiple comments have been added for a patient, then the comments are displayed in reverse chronological order in the Comments pane. Only the last comment added is displayed in the Last Comment field.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. If you are an Administrator user, then you can set or modify the permissions for a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See "To set or modify the Read/Write permissions for a patient record" on page 98.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Patient tab, Patient sub-tab, Related Patients pane

The Related Patients pane on the Patient sub-tab shows all the current relationships for the patient, and the last comment that was added for each related patient. You can double-click a patient on the Related Patients tab to display the information for the related patient in a new Patient tab. The currently opened Patient tab remains open, but the focus is changed to the new Patient tab. You can right-click in the pane to open a context menu with the following options:

Table 5-4: Patient tab, Patient sub-tab, Related Patients pane context menu

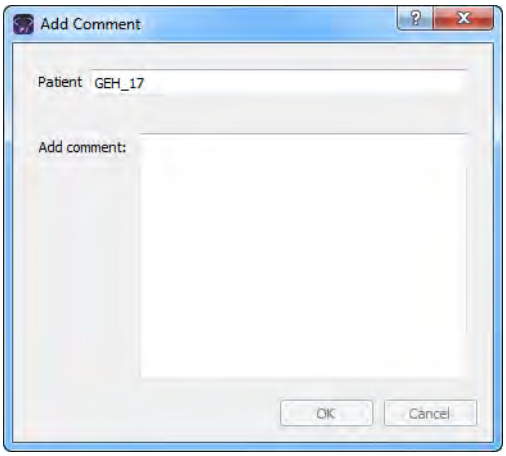
Option	Description
Reload in Current Tab	Displays the information for the selected related patient in the currently opened Patient tab.
Open in New Tab	Displays the information for the selected related patient in a new Patient tab. The currently opened Patient tab remains open, but the focus is changed to a new Patient tab that displays the information for the related patient.
Add New Relationships	Opens the Add New Patient dialog box so that you can add a relationship to the selected related patient. By default, the Is Related to Existing Patient option is selected and unavailable. Repeat Step 3 of "To add new patients one at a time" on page 118 as many times as needed to add all the necessary patient relationships to the related patient.
Remove Patient's Relationships	Removes all the relationships that have been added for the patient.
Add All to Custom Report	Selects all the data that is displayed for all the related patients for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Add Comment	Opens the Add Comment dialog box. You add a comment for the selected related patient in the Add Comment field. The comment is displayed in the Last Comment field in the pane. *Figure 5-6: Add Patient Comment dialog box*  **Note:** If multiple comments have been added for a related patient, then only the last comment added is displayed in the Last Comment field. To see all the comments that have been added for a related patient, you must open the related patient in a new Patient tab.

Table 5-4: Patient tab, Patient sub-tab, Related Patients pane context menu (Continued)

Option	Description
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. If you are an Administrator user, then you can set or modify the permissions for a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See "To set or modify the Read/Write permissions for a patient record" on page 98.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Patient tab, Patient sub-tab, Comments pane

The Comments pane on the Patient sub-tab shows all the comments that have been added for the patient in reverse chronological order. You can right-click in the pane to open a context menu with the following options:

Table 5-5: Patient tab, Patient sub-tab, Comments pane context menu

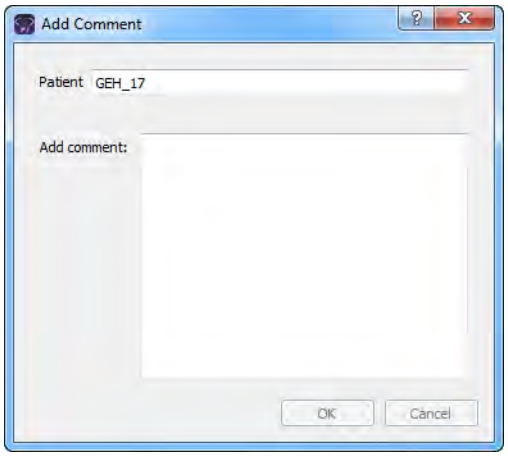
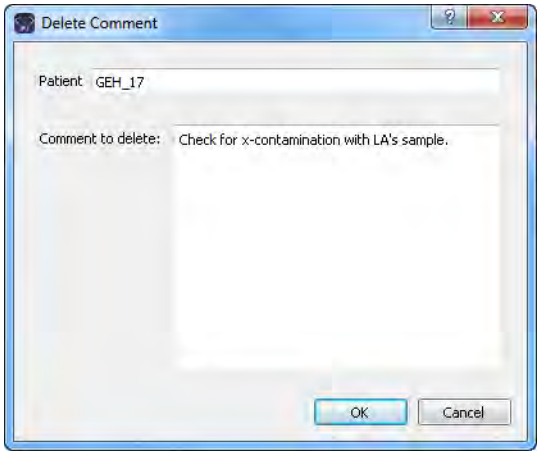
Option	Description
Add Comment	Opens the Add Comment dialog box. You add a comment for the selected patient in the Add Comment field. The Comments are displayed in reverse chronological order in the pane. *Figure 5-7: Add Patient Comment dialog box* 

Table 5-5: Patient tab, Patient sub-tab, Comments pane context menu (Continued)

Option	Description
Delete Comment	Opens the Delete Comment dialog box. Click OK to delete the comment for the sample and close the dialog box. *Figure 5-8: Delete Comment dialog box* 
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

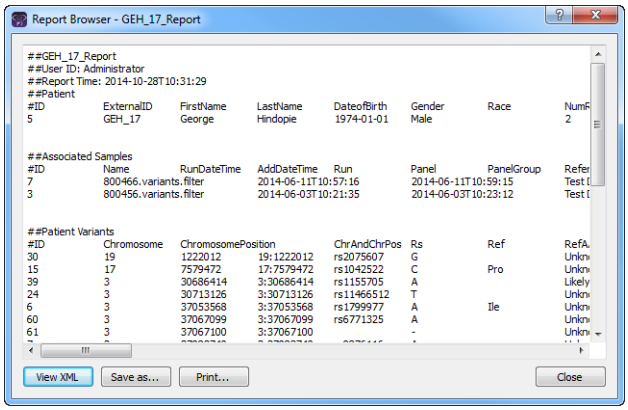
Patient tab, Patient sub-tab, Reports pane

The Reports pane on the Patient sub-tab lists all the reports that have been generated for the selected patient and saved to the Geneticist Assistant database.

You can also select Reports > Saved Reports on the Geneticist Assistant main menu to open a (Patient) Reports tab that displays these same reports.

You can right-click a report entry to open a context menu with the following options:

Table 5-6: Patient tab, Patient sub-tab,. Reports pane context menu

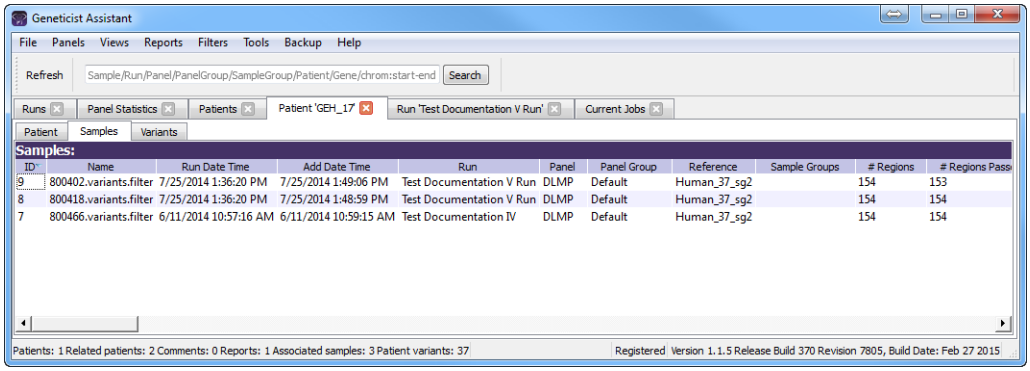
Option	Description
View Report	Opens the Report Browser dialog box, which provides an onscreen preview (in plain text) of the selected report. **Note:** You can also double-click the report entry to open the Report Browser dialog box. *Figure 5-9: Report Browser dialog box*  The following options are available for the report: • View XML - Toggles the onscreen display of the report between XML and plain text. • Save As - Saves the report as a .csv file. By default, the report is named as <External ID>.csv but you can always change the name and you can also select the location in which to save the report. • Print—Prints the onscreen report to a printer of your choice.
Delete Report	Deletes the selected report from the Geneticist Assistant database.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

For detailed information about generating and saving both standard and custom Patient reports, see Chapter 7, "Geneticist Assistant Reports," on page 203.

Patient tab, Samples sub-tab

The Samples sub-tab on the Patient tab has a single pane, the Samples pane, that displays all the samples that are associated with the patient as well as information about the sample runs.

Figure 5-10: Patient tab, Samples sub-tab

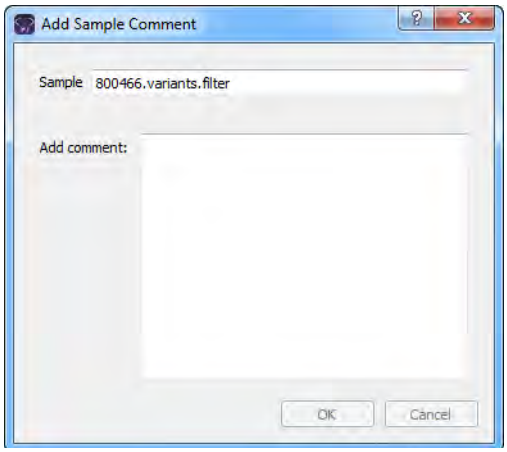


You can right-click in the pane to open a context menu with the following options:

Table 5-7: Patient tab, Samples sub-tab, Samples pane context menu

Option	Description
Sample Details	Opens the Sample tab, which contains detailed information about the associated sample, including the variant list. See “The Sample Tab” on page 173.
Coverage QC	Opens the Coverage QC tab, which displays the coverage information for the associated sample. See “Coverage QC tab (via the Samples pane on the Run tab)” on page 144.
Coverage Regions	Opens the Coverage Regions tab, which lists the coverage information for every region in the panel for the associated sample. See “Coverage Regions sub-tab (Coverage QC tab)” on page 146.
Delete Sample	Deletes the association between the sample and the patient and removes the sample entry from the Samples pane.
Add All to Custom Report	Selects all the data that is displayed for all the associated samples in the run for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.
Create Batch Variants Report	Generates the Variant report for all the selected variants in the selected associated sample. (Press and hold the CTRL to select multiple samples.) See “To manage a Batch Variants report (Samples sub-tab on the Patient tab)” on page 108.

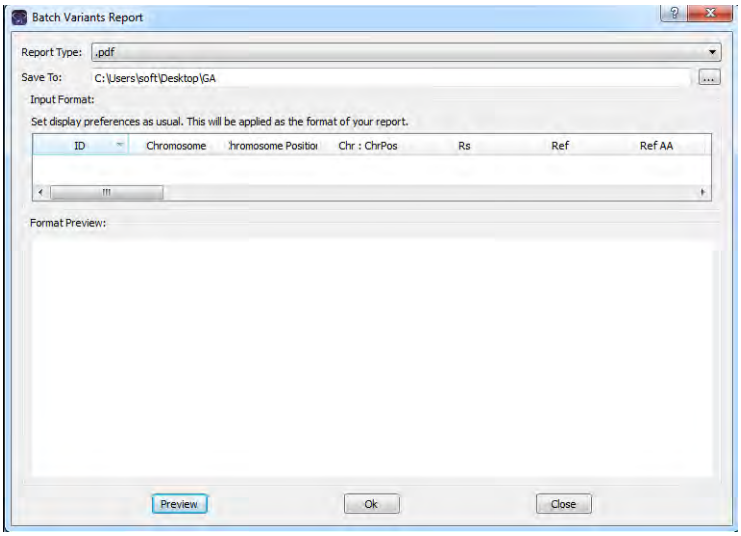
Table 5-7: Patient tab, Samples sub-tab, Samples pane context menu (Continued)

Option	Description
Add Comment	Opens the Add Sample Comment dialog box. You add a comment for the associated sample in the Add Comment field. The comment is displayed in the Comments field in the Samples pane. *Figure 5-11: Add Sample Comment dialog box*  **Note:** If multiple comments have been added for a sample, then these comments are displayed in reverse chronological order in the Comments pane on the Sample Data sub-tab for the Sample tab. Only the last added comment is displayed in the Comments field in the Samples pane on the Samples data sub-tab on the Patient tab and in the Samples pane on the Run tab.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the Patient tab. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

To manage a Batch Variants report (Samples sub-tab on the Patient tab)

A *Batch Variants report* is a report that includes information for variants over a specified date range or other parameters. A *Batch Variants report configuration* determines the data that is to be included in the report, and if the report is to be available only to the user who created it, or to all users. The configuration does not include the report format (PDF, HTML and so on), or the location in which the report is to be saved. Instead, you must specify the format and location in which to save the report every time you generate a report for a new or saved configuration.

Figure 5-12: Batch Variants Report dialog box



You can generate a Batch Variants report according to a new or saved configuration, modify a Batch Variants report configuration, or delete a Batch Variants report configuration. See:

- "To generate a Batch Variants report based on a new configuration" below.
- "To generate a Batch Variants report based on a saved configuration" on page 109.
- "To modify a Batch Variants report configuration" on page 110.
- "To delete a Batch Variants Report configuration" on page 111.

To generate a Batch Variants report based on a new configuration

When you generate a Batch Variants report, the report name is automatically assigned based on the variant ID for which you are generating the report (for example, MSH2_GC) and you cannot change this. You must select a location in which to save the report.

1. On the Batch Variants Report dialog box, on the Report Type dropdown list, select the report type. (The default value is PDF.)

2. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
   - Leave this location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Optionally, to view an onscreen preview of the report before it is generated and saved, click Preview.
4. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.
5. Click OK.

   The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

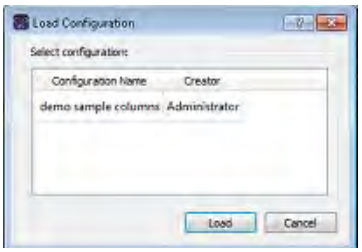
To generate a Batch Variants report based on a saved configuration

1. On the Batch Variants Report dialog box, select the output format for the report.
2. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
   - Leave the location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.
4. Click Load Configuration.

   The Load Configuration dialog box opens. The dialog box displays all your private and public configurations as well as all the configurations that other users have made public.

Figure 5-13: Load Configuration dialog box



5. Select the configuration that you are loading, and then click Load.

   The Load Configuration dialog box closes. The Select Columns dialog box remains open. The pane display is updated dynamically after you load the configuration.

6. Select a configuration, and then click Load.

   The Load Configuration dialog box closes. The Select Columns dialog box remains open. The dialog box now reflects the columns that are to be included in the report based on the selected configuration.

7. Click Close (x) to close the Select Columns dialog box.

8. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.

9. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.

10. Click OK.

    The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To modify a Batch Variants report configuration

When you modify a Batch Variants report configuration, any reports that were generated according to the “old” (previous) configuration are not affected. Going forward, the report is generated based on the “new” configuration.

1. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank.

2. Do one of the following:

   - Leave this location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button  , and then browse to and select the location in which the report is to be saved.

3. Right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.

4. To modify the report configuration, see “To create/modify a pane configuration” on page 32.

5. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.

6. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box opens, with a message that indicates where the report was saved.

7. Click OK.

   The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To delete a Batch Variants Report configuration

You can delete a Batch Variants report configuration only if you created the report. When you delete a Batch Variants report configuration, any reports that were generated according to the "old" (previous) configuration are not affected. Going forward, the "old" configuration is simply not available for selection.

You are not prompted about confirming the deletion of a Batch Variants report configuration, so be very sure that the deletion must be deleted before doing so.

1. On the Batch Variants Report dialog box, right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.

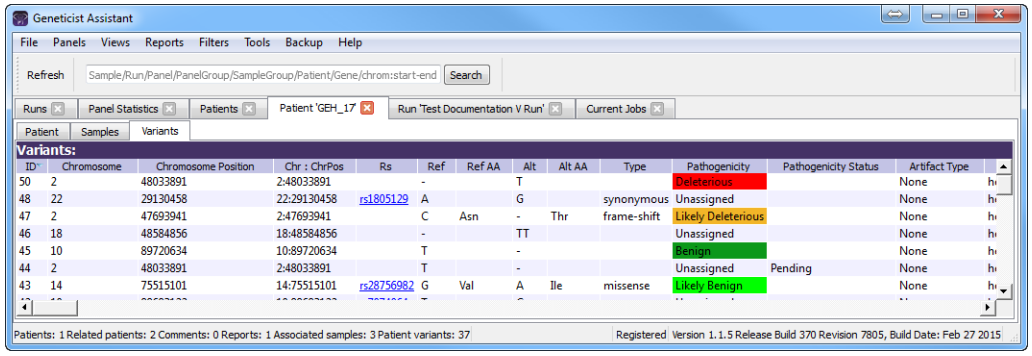
2. See "To delete a pane configuration" on page 34.

3. Click Close (x) to close the Batch Variants Report dialog box.

Patient tab, Variants sub-tab

The Variants sub-tab on the Patient tab has a single pane, the Variants pane, that displays all the variants in all the samples that are associated with the patient. With the exception of a few omitted columns, the information that is displayed in the Variants pane is identical to the information that is displayed, by default, for a variant on the Variants sub-tab on the Coverage QC tab. (See "Variants sub-tab (Coverage QC tab)" on page 148.)

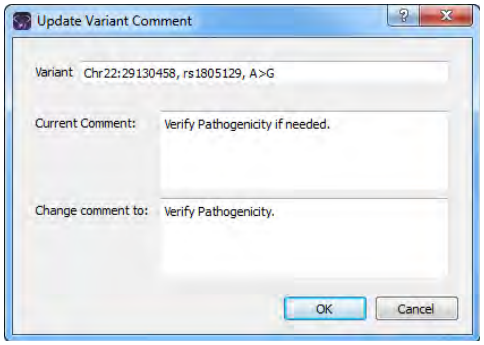
Figure 5-14: Patient tab, Variants sub-tab



You can do the following for a variant that is displayed on the sub-tab:

- You can specify how the information about the variants is to displayed on this sub-tab. See "To specify variant preferences" on page 53.
- You can select a different transcript for the variant on the Transcript dropdown list.
- You can single click twice in the Variant Comment field to select the current comment in the field and edit it, or if the field is blank, enter a new comment. After you add or edit a comment, and then click out of the field, the Update Variant Comment dialog box opens.

Figure 5-15: Update Variant Comment dialog box



The dialog box displays the variant for which you are adding or editing the comment, the current (old) comment, and the new comment. You can make additional edits to the new comment needed, and then click OK to close the dialog box and save the new or modified comment for the variant.

You can also use the Add Variant Comment option that is available on the context menu for the variant to add a new variant comment. (See "Add Variant Comment" on page 114.)

- You can click once in the Type field, the Zygosity field, the HGVS Genomic field, the HGVS Coding field, the HGVS Protein field, and the Read Balance field to select the current value in the field and edit it, or if the field is blank, enter a new value.

If you make a change to any of these values for the variant, then these changes are recorded for the variant on the Variant Changes pane on the Variant tab. See "Variant Changes pane" on page 196. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in the Geneticist Assistant application.

- You can right-click a variant in the pane to open a context menu with the following options:

Table 5-8: Patient tab, Variants sub-tab, Variants pane context menu

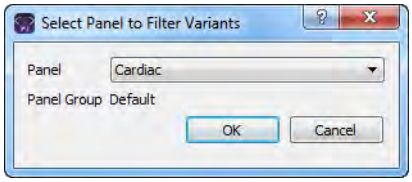
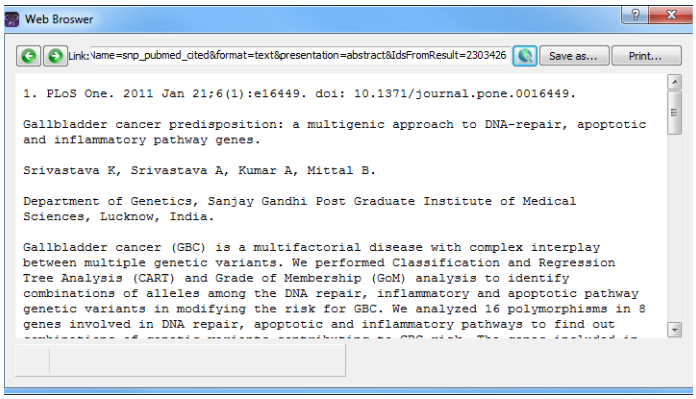
Option	Description
Variant Details	Opens the Variant tab for the selected variant. See "The Variant Tab" on page 190.
Show Variants Filtered by Panel	Displays all the variants in all the associated samples for the selected patient filtered by a selected panel. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected patient. *Figure 5-16: Select Panel to Filter Variants dialog box* 
View in Alamut	Opens the variant for viewing in Alamut. **Note:** To view a variant in Alamut, your Alamut settings must be specified. See "To specify Alamut settings" on page 54.
View PubMed Abstract	Searches PubMed for the variant and displays the appropriate abstract in the PubMed window. *Figure 5-17: PubMed window*  You can do the following for the abstract: • In the Link field, enter a different PubMed URL, and then click the Go To icon to navigate to abstract. • Click Save As to save the abstract as a PDF with a name and in a location of your choosing. • Click Print to print the abstract.
Add Selected Variants to Custom Report	Selects all the data that is displayed for the selected variant for inclusion in a custom report. Press and hold the CTRL key to select multiple variants. See Chapter 7, "Geneticist Assistant Reports," on page 203.

Table 5-8: Patient tab, Variants sub-tab, Variants pane context menu (Continued)

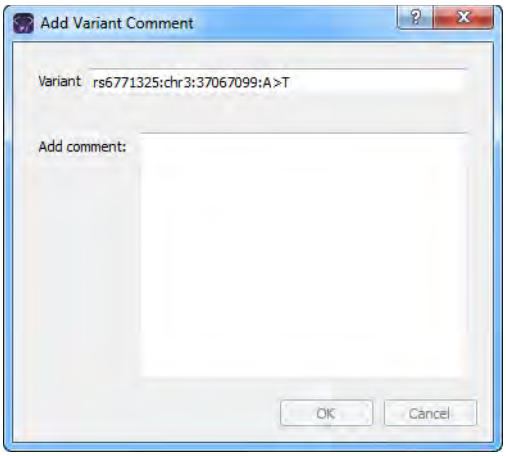
Option	Description
Add All Variants to Custom Report	Selects all data that is displayed for all the variants for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.
Export Selected Variants to VCF	Exports the selected variants to a VCF file. Press and hold the CTRL key to select multiple variants. You must specify both the file name and the location in which to save the file.
Export All Variants to VCF	Exports all the variants to a VCF file. You must specify both the file name and the location in which to save the file.
Add Variant Comment	Opens the Add Variant Comment dialog box. You can add a comment for the variant in the Add Comment field, and then click OK. The comment is displayed in the Variant Comment field in the Variants pane on the Search Results tab, the Variant Comment field on the Variants sub-tab on the Patient tab, the Variant Comment field on the Variants sub-tab on the Coverage QC tab, the Sample Variant Data sub-tab on the Sample tab, the Variant Comment field in the Variant pane on the Variant tab, and the Variant Comments pane on the Variant tab. *Figure 5-18: Add Variant Comment dialog box*  **Note:** If multiple comments have been added for a variant, then with the exception of the Variant Comments pane on the Variant tab, only the last added comment is displayed in all the locations that are detailed above. All the comments that have been added for a variant, including the last added, are displayed in reverse chronological order in the Variant Comments pane on the Variant tab.

Table 5-8: Patient tab, Variants sub-tab, Variants pane context menu (Continued)

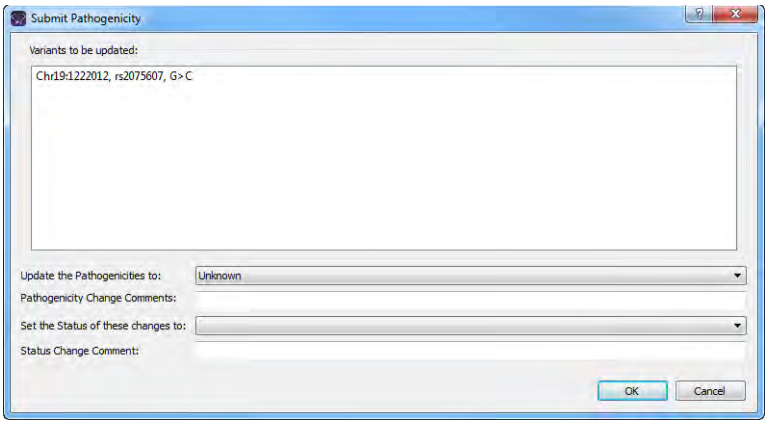
Option	Description
Update Pathogenicity	Opens the Submit Pathogenicity dialog box. You can update the pathogenicity and/or pathogenicity status for the selected variant in this dialog box. You can also add a comment for the variant. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be. After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant. *Figure 5-19: Submit Pathogenicity dialog box*  **Note:** You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box. You also press and hold the CTRL key to select multiple variants for updating the pathogenicities and the pathogenicity statuses at the same time. **Note:** You can also update the Pathogenicity and the Pathogenicity Status for a variant on the Variants sub-tab on the Coverage QC tab and the Sample Variant Data sub-tab on the Sample tab. The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See "Managing Objects" on page 41. • Optionally, you can add supporting information for this pathogenicity change. See "To add supporting information for a pathogenicity change" on page 198. • You always have the option of updating just the pathogenicity status for a variant at a later date. See "Update Pathogenicity Status" below.

Table 5-8: Patient tab, Variants sub-tab, Variants pane context menu (Continued)

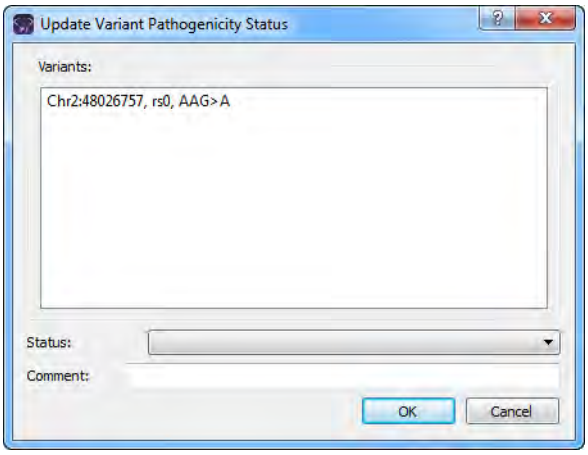
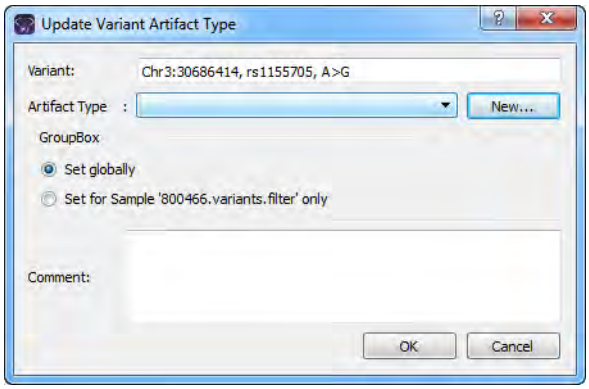
Option	Description
Update Pathogenicity Status	Opens the Update Variant Pathogenicity Status dialog box. You can update the pathogenicity status for the selected variant in this dialog box. You can also add a comment for the variant. *Figure 5-20: Update Variant Pathogenicity Status dialog box*  The following information is relevant to updating the pathogenicity status for a variant: • You can also double-click in the Pathogenicity Status column to open the Update Variant Pathogenicity Status dialog box. • You can press and hold the CTRL key to select multiple variants for updating the pathogenicity statuses at the same time. • You can also update the Pathogenicity Status for a variant on the Variants sub-tab on the Coverage QC tab and the Sample Variant Data sub-tab on the Sample tab. • By default, only two statuses, Pending or Confirmed, are available. You have the option of adding custom pathogenicity statuses. See “Managing Objects” on page 41.

Table 5-8: Patient tab, Variants sub-tab, Variants pane context menu (Continued)

Option	Description
Update Artifact Type	Opens the Update Artifact Type dialog box. You specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type. *Figure 5-21: Update Variant Artifact Type dialog box*  When you specify the artifact type, you can set the artifact type as the following: • Set globally—Selected by default. Set this artifact type for the variant, regardless of the sample in which the variant is detected. • Set for Sample—Set this artifact type for this variant only for the indicated sample. You also have the option of adding comments for the variant artifact type. The following information is relevant to updating the artifact type for a variant: • You can press and hold the CTRL key to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant in the Variants pane on the Variants sub-tab on the Coverage QC tab, on the Variants sub-tab on the Panel Statistics tab via the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See "Managing Objects" on page 41.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations. **Note:** If you set column display preferences here, then you can apply these same display preferences for variants in all the variant sub-tabs and panes (with the exception of the Sample Group columns on the Sample Variant Data sub-tab on the Sample tab and the Variant pane on the Variant tab) in a single step. See "To apply shared preferences or to clear all tab preferences" on page 51.

Adding Patients

In addition to managing settings (chemistries, instruments, and so on) for your runs, you can manage the patients for your runs. A *patient* refers to an individual from whom a sample was taken. Managing patients for your runs consists of adding new patients to the Geneticist Assistant database. When you add new patients, you can manually add patients one at a time, or you can use the a batch import process to add multiple patients in a single step. Moreover, if your VCF or BAM sample file names begin or end with the patient ID, then Geneticist Assistant can automatically associate the appropriate sample with the patient. If you are loading a standalone VCF file, then just the VCF file name must begin or end with the patient ID. If you are loading both a VCF file and a BAM file, then the names for both files must begin and/or end with the patient ID.

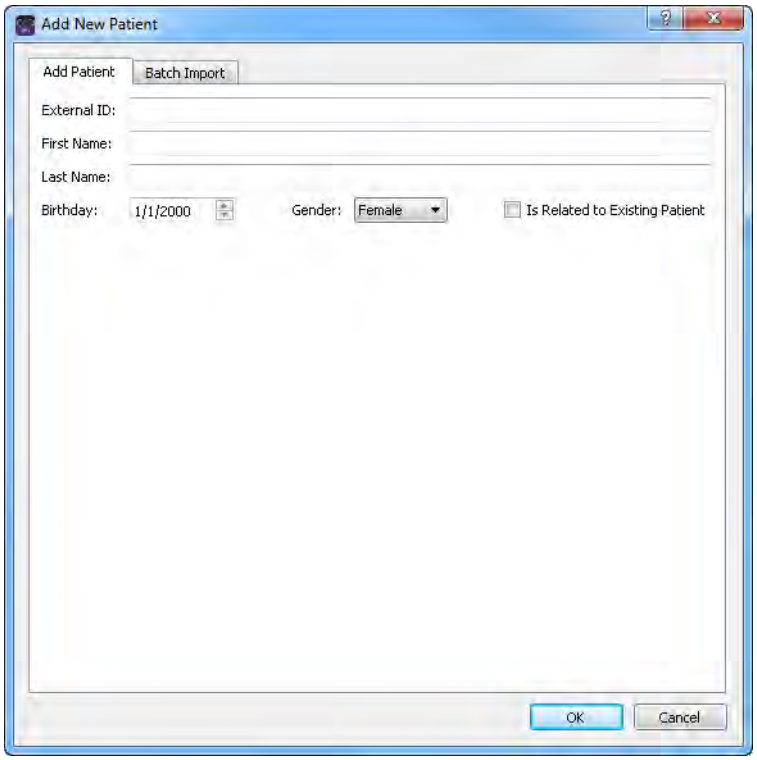
To add new patients one at a time

When you add a patient one at a time, you can specify the name, birthdate, and gender for the patient as well as a unique ID for the patient. You can also add relationships to the patient.

1. On the Geneticist Assistant main menu, click File > Add Patient.

   The Add New Patient dialog box opens. The Add Patient tab is the opened tab.

Figure 5-22: Add New Patient dialog box, Add Patient tab



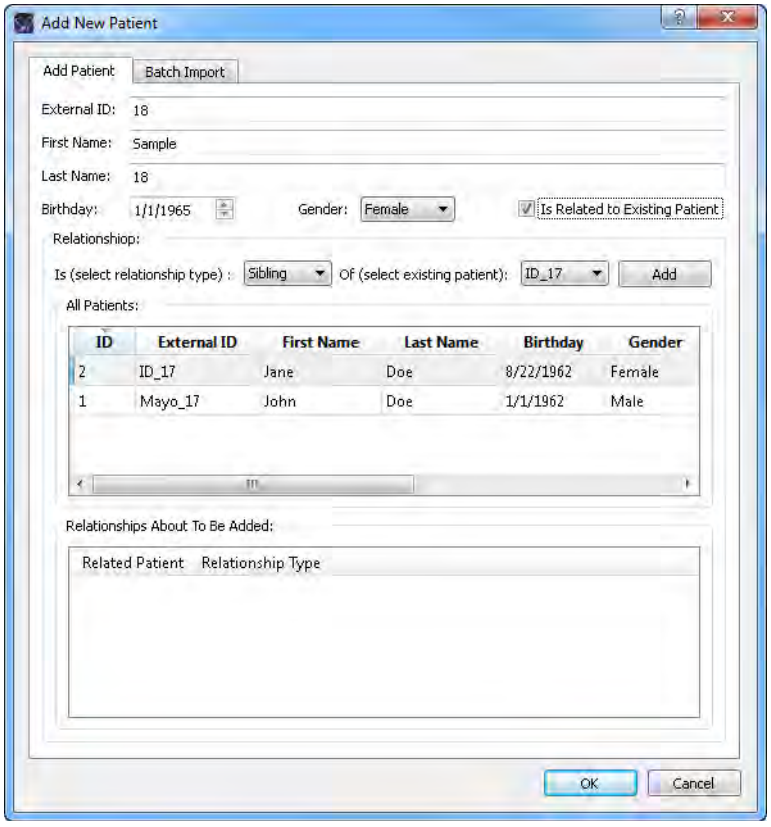
2. Enter the information for the new patient.

Option	Description
External ID	A unique ID for the patient.
First Name	The first name for the patient.
Last Name	The last name for the patient.
Birthday	The patient's birthday. You must select each value individually, and then you can manually enter a new value, or you can use the Up/Down arrows.
Gender	Male or Female.

3. If applicable, to specify relationships for the new patient, select Is Related to Existing Patient and then do the following:
   - Select the relationship type (Sibling (the default value), Child, or Parent).
   - Select the existing patient for this relationship. (Select the patient on the select existing patient list, or double-click the patient entry in the All Patients pane.)
   - Click Add.

   The Relationships About to Be Added list display the relationship that you have specified for the new patient.

Figure 5-23: Add New Patient dialog box, Relationship options



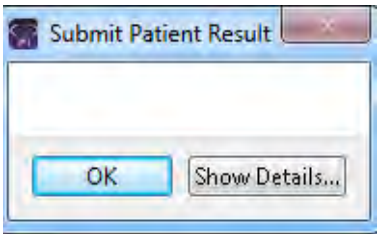
4. Repeat Step 3 to add as many relationships for a patient as needed.

If you add a relationship in error, right-click the relationship, and then select Remove Selected Rows. Press and hold the CTRL key to select multiple relationships for deletion.

5. Click OK.

   The Add New Patient dialog box closes. The Submit Patient Result dialog box opens.

Figure 5-24: Submit Patient dialog box



6. Optionally, click Show Details to view the details of all the relationships that you added for the new patient and then click OK to close the dialog box; otherwise, click OK to close the dialog box immediately.

To add new patients with the batch import process

You use a batch import process to add multiple patients in a single step to the Geneticist Assistant database. The batch import file must be either a comma-separated values (.csv) file, a tab-delimited text (.txt) file, or a tab-separated values (.tsv) file. The file must contain the following columns of information—the patient ID, the patient last name, the patient first name, the patient date of birth, the patient gender, the patient race, and the patient relationships. Figure 5-25 below shows the standard column order and the standard column names. If your import file does not follow either the standard column order or the standard column names, then you must manually indicate the order of the columns in your file. The patient relationship information requires two columns. The first column contains the ID of the related patient. The second column contains the relationship type (for example, Sibling). You must always indicate the location of the first relationship column.

Figure 5-25: Standard column order and names

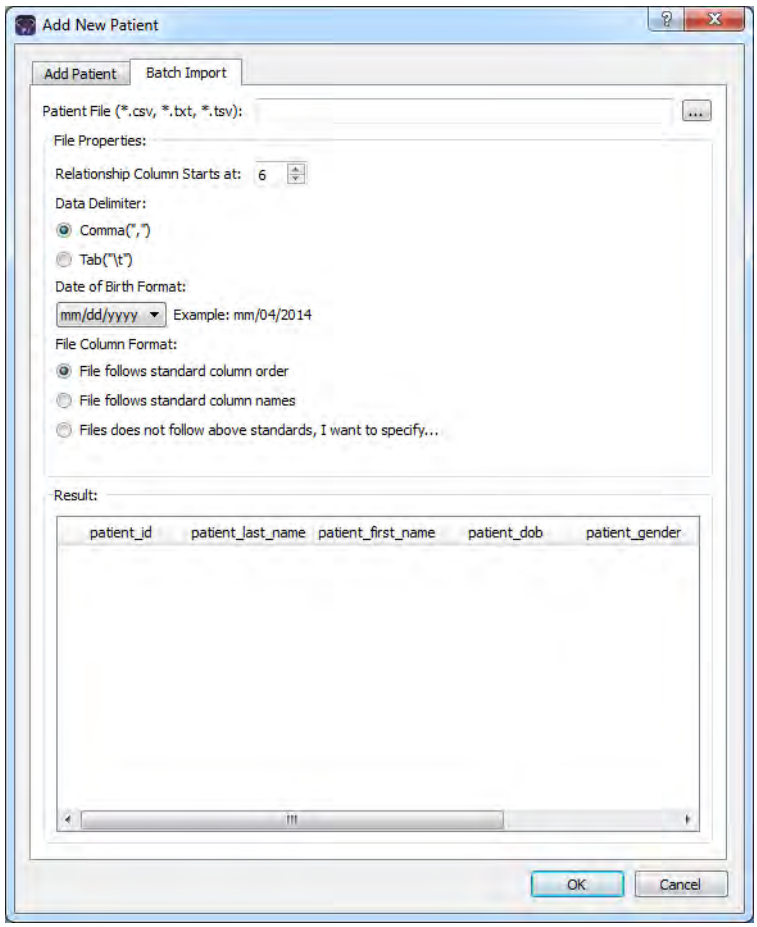
patient_id	patient_last_name	patient_first_name	patient_dob	patient_gender	patient_race	patient_relationships
BC-13-20683	Doe	Jane	1920/3/2	Female	Causcasian	BC-13-20476-Spouse;
BC-13-15487	Doe	Jill	1946/6/10	Female	Causcasian	BC-13-20476-Father;
BC-13-15487	Doe	Jill	1946/6/10	Female	Causcasian	BC-13-20683-Mother;

1. On the Geneticist Assistant main menu, click File > Add Patient.

   The Add New Patient dialog box opens. The Add Patient tab is the opened tab. See Figure 5-22 on page 118.

2. Click the Batch Import tab to open it.

Figure 5-26: Add New Patient dialog box, Batch Import tab



3. Next to the Patient File field, click the Browse button ... to open the Select file dialog box, and then browse to and select your patient file.
4. Specify the properties of your patient file.

Property	Description
Relationship Column Starts at	Indicate where the Relationship column starts. (The default value is Column 6.)
Data Delimiter	Indicate the delimiter used (Comma or Tab).
Date of Birth Format	Select the format used for the patients' birthdays.
File Column Format	Select one of the following to indicate the file format. • File follows standard column order. • File follows standard column names. • File does not follow above standards. I want to specify.

5. If your file follows the standard column order or the standard column names, go to Step 8; otherwise, if your file does *not* follow either of these standards, go to Step 6.
6. In the Result pane, rearrange the column headers so that they are in the same order as the column headers in your patient file.

For information about rearranging columns, see "Rearrange the order of data columns" on page 35.

7. Click Check With File.
   - The file is loaded and the patient information is displayed in the Result pane. If your file format is correct, go to Step 8.
   - If your file format is incorrect then, you must click Incorrect, Redo, and then make the necessary changes to the column headers. Click Check With File again to verify that your file has the correct format. Only after you verify that your file has the correct format can you continue to Step 8.
8. Click OK.

   The Add New Patient dialog box closes. The patients are imported into your Geneticist Assistant database.

Adding a Run

A *run* refers to loading a BAM or DAT file *and* a VCF file or just a VCF file into Geneticist Assistant. for the purposes of importing a list of variants into the Geneticist Assistant database and/or reviewing a list of variants. When you add a run, you must specify the run and sample settings, which include the chemistry, the instrument, the reference, the panel, and the panel group. You must also select the files that are to be loaded. Optionally, you can choose to modify the run date/time and/or assign the run to a sample group. You specify these settings on a per run basis, or you specify these settings on a per sample basis. At a minimum, you must always load a VCF file. Optionally, you can also load a BAM or a DAT file to include coverage information for the panel regions.

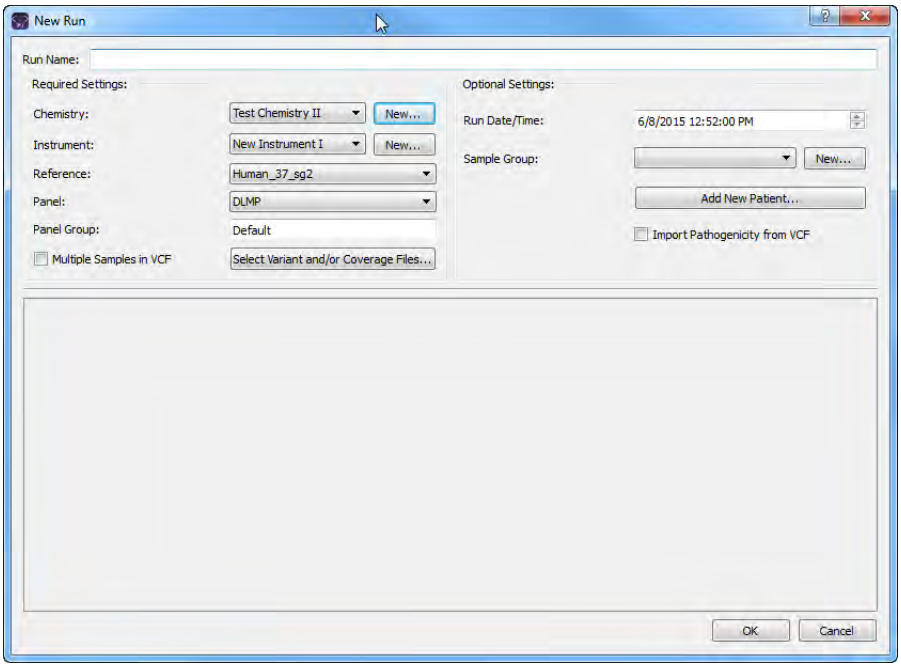
The DAT file is the coverage file that is produced by NextGENe. The file is always named Coverage2DInfo_Wrap.dat and you cannot change this. The Coverage2DInfo_Wrap.dat file loads faster than a BAM file; however, if you load this DAT file instead of a BAM file, then pileup information is not available on the Coverage QC tab. (See "Read Pileups pane" on page 145.) The Coverage2DInfo_Wrap.dat file is located in the output folder for the NextGENe analysis: <projectname>\<projectname>files\sample.

To add a run

1. On the Geneticist Assistant main menu, click File > New Run.

   The New Run dialog box opens.

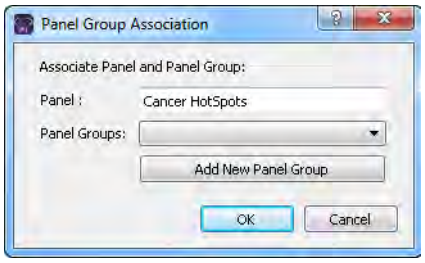
Figure 5-27: New Run dialog box



2. Enter the name for the new run.

The name can have a maximum of 255 characters. Spaces are allowed.

3. Specify the run settings. Note the following about these settings:

Setting	Description
Required Settings	
Chemistry	If the correct chemistry is not available, then you can add it. Click New to open the Add New Chemistry dialog box and add the new chemistry.
Instrument	If the correct instrument is not available, then you can add it. Click New to open the Add New Instrument dialog box and add the new instrument.
Reference Files	If the correct reference files are not available, then you might need to specify the reference directory and/or add the necessary reference files to the directory. (See "Specifying your Geneticist Assistant Settings" on page 45.) **Note:** SoftGenetics provides the necessary references files for your Geneticist Assistant installation. Contact SoftGenetics about downloading these files. **Note:** This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
Panel	If the correct panel is not available, then you might need to add the necessary panel. See "Adding a Panel" on page 79. After you select a panel, two results are possible: • If the selected panel has previously been assigned to a panel group, then panel group is automatically selected and the name is displayed in the Panel Group field. • If the selected panel has *not* been previously assigned to a panel group, then the Panel Group Association dialog box opens and you must select the appropriate panel group. *Figure 5-28: Panel Group Association dialog box*  **Note:** This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
Multiple Samples in VCF	Select this option if you are loading a VCF file that contains variants from multiple samples.

Setting	Description
Optional Settings	
Run Date/Time	The default value for the Run Date and Time is the current day's date and time, but you can change one or both values as needed. To change the value, select the value that is to be changed, for example, the month, and then you can manually enter the new value, or you can use the Up/Down field arrows to change the value. **Note:** You must select each value that is to be changed one at a time. If you set the Run Date/Time here, then this is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
Sample Group	Optional. Used for a group of samples that are to be processed repeatedly over a period of time, for example, a group of control samples for which the coverage or variant frequency is to be tracked. If the correct Sample Group is not available, then you can add it. Click New to open the Add New Sample Group dialog box and add the new sample group. **Note:** This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
Import Pathogenicity from VCF	Select this option if you are loading a VCF file that contains pathogenicity information and you want to import this information into the run results. **Note:** This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.

4. To load your files, do the following:

   - Click Select Variant and/or Coverage Files to open the Select Files dialog box.
   - In the Select Files dialog box, browse to and select the coverage (BAM or DAT) files and/or variant (VCF) files that are to be loaded, and then click Open.

   When selecting the files that are to be loaded, note the following:

   - You can load a VCF, BAM, or DAT file for a new run that has been submitted previously for another run. You can keep the sample name the same, or you can modify the sample name as needed.
   - You can use the same run name to load additional sample files (VCF, BAM and/or DAT) for a previously submitted run. The additional sample files are processed and appended to the run.
   - At a minimum, you must always load a VCF file. Optionally, you can also load a BAM or a DAT file to include coverage information for the panel regions.
   - If you have a *single* sample to load, and the VCF and BAM or DAT file names do *not* contain corresponding fields in the file names, then select the VCF, BAM or DAT file, and then after you click Open, the file is placed in the appropriate field for the sample entry. You can then click the Browse button  next to the appropriate file type to load the corresponding file for the sample. For example, if you loaded a

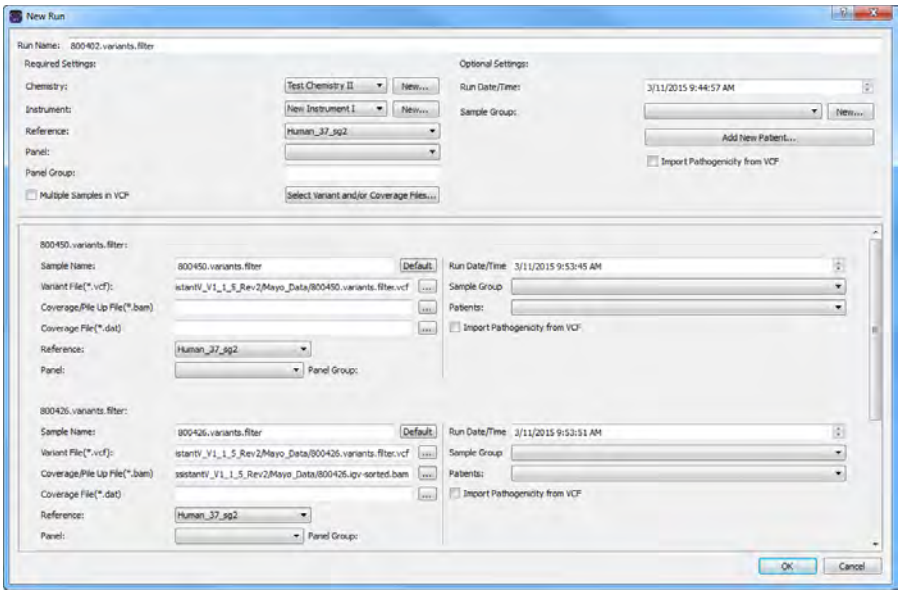
BAM or DAT file, then you can click the Browse button … next to the Variant File(*.vcf) field for the sample entry to load the needed VCF file for the sample.

- If you have multiple samples to load and the VCF and BAM or DAT files do *not* contain corresponding fields in the file names, then press and hold the CTRL key as you select only the VCF files, BAM files, or DAT files, and then after you click Open, each file is placed in its own sample entry. For each sample entry, you can then click the Browse button … next to the appropriate file type to load the corresponding file. For example, if you loaded a BAM or DAT file, then you can click the Browse button … next to the Variant File(*.vcf) field for the sample entry to load the needed VCF file for the sample.
- If you are loading both a VCF file and a BAM or DAT file, and the files *do* contain corresponding fields in the file names, then press and hold the CTRL key as you select both the VCF and BAM or DAT files, and then after you click Open, both files are loaded into the appropriate fields for the *same* sample entry in a single step. You can load a single VCF/BAM file or VCF/DAT combination this way, or you can load multiple VCF/BAM file or VCF/DAT file combinations. Each correspondingly named VCF/BAM file or VCF/DAT file combination is placed in its own sample entry.
- If you have multiple BAM files for a sample, you do not need to submit each file as an individual sample, and then assign each file to the same patient. Instead, you can use the Merge BAM Files tool to merge all the BAM files into one file and then submit the file as a single sample. See "Merging BAM Files with the Merge BAM Files Tool" on page 222.
- By default, the sample name is set to the VCF file name. The name is displayed at the top of the sample entry. You can modify the sample name.
- If you load a sample file in error, then you can right-click the sample name, and then click Remove this sample.

After the files are loaded for a sample, the full directory paths (directory and file name) for the loaded files are displayed in the appropriate fields for each sample entry at the bottom of the New Run dialog box. See Figure 5-29 on page 127.

After you load sample files from a specific directory, these files must remain in this directory. You cannot move sample files to a different directory after you have successfully submitted the files for a run.

Figure 5-29: New Run dialog box with loaded sample files



5. Optionally, for each sample, do any or all of the following as needed:

   - Modify the sample name.

You can click Default at any time to return the sample name to its default value.

   - By default, the Reference, Panel, Run Date/Time, Sample Group, and Import Pathogenicity from VCF values that are indicated for a sample are the run-specific values that you specified in Step 3. You can modify any or all these values on a per sample basis.

If you modify the panel on per sample basis, then the panel group might also be modified, but you cannot modify the panel group directly.

   - To associate a patient with a sample, select a patient on the Patients dropdown list.

If the correct patient is not available, then you can click Add New Patient to add the patient. See "Adding Patients" on page 118.

6. Click OK.

   The Current Jobs tab opens and a progress bar that shows the progress of loading the files is displayed on the Current Jobs tab. After the files are successfully loaded, the progress bar is removed from the Current Jobs tab. You return to the Runs tab. The new run is displayed on the Runs tab. You can now review the data for the run. See Chapter 6, "Geneticist Assistant Data Review," on page 129.

Chapter 5
Managing Patients and Runs

Chapter 6
Geneticist Assistant Data Review

Many options are available for reviewing your data in Geneticist Assistant.

This chapter covers the following topics:

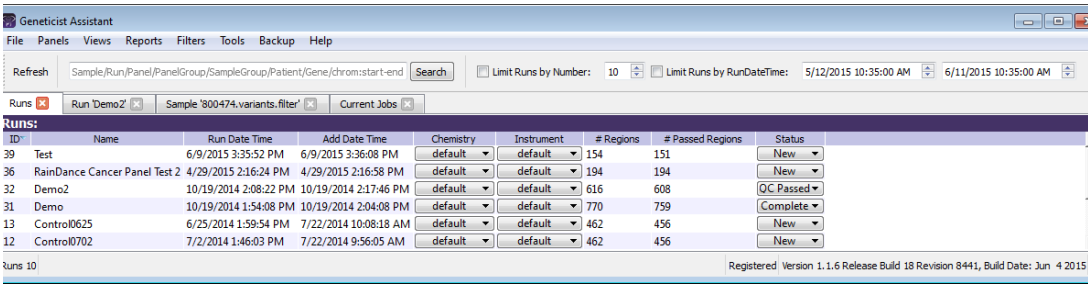
Chapter 6
Geneticist Assistant Data Review

The Runs Tab

When the Geneticist Assistant main window first opens, the Runs tab is displayed in the Geneticist Assistant main window. The Runs tab has a single pane, the Runs pane, that displays information about the runs that have been carried out in your Geneticist Assistant installation.

- If you are an Administrator user, then the Runs pane can display all the runs submitted by all users to your Geneticist Assistant database. By default, when the tab first opens, only up to the 10 most recently submitted runs are displayed and the runs are sorted based on the Run ID.
- If you are a standard user, then the Runs pane can display all the runs that you have submitted to your Geneticist Assistant database as well as all the runs that have been submitted by other users and for which you have been granted at least the Can Read permission. By default, when the tab first opens, only up to the 10 most recently added runs that fit one of these two criteria are displayed and the runs are sorted based on the Run ID.

Figure 6-1: Runs tab

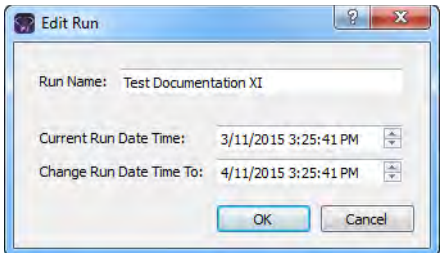


You can do the following on the Runs tab:

- Limit the runs that are displayed by number. (The default value is ten.)
- Limit the runs that are displayed by a range for the run date and time. (The default value is the last 30 days, with the ending date/timestamp set to the current day's date and time.)
- Click in any of the following fields—Name or Run Date/Time—to select the current value, and modify it as necessary, and then click out of the field.
- Select a different Chemistry and/or Instrument for the run.

After you modify the value for Name or Run Date/Time and click out of the field, or select a different chemistry or instrument for the run, an Edit Run dialog box opens. The dialog box displays the name of the run for which you are editing a value, the current (old) run value, and the new run value. You can make any further changes to the new run value as needed, and then click OK to close the dialog box, and save the new value for the run. See *Figure 6-2 on page 132.*

Figure 6-2: Edit Run dialog box



- Set the status for the run. When a run is first submitted, the status is set to New. You can change the status from New to one of three default values (QC Passed, Reviewed, or Complete) for the run, or if custom values have been added for the run, then you can select from one of these values. (See "Managing Objects" on page 41.)
- Right-click a run entry to open a context menu with the following options:

Table 6-1: Runs tab, Runs pane context menu

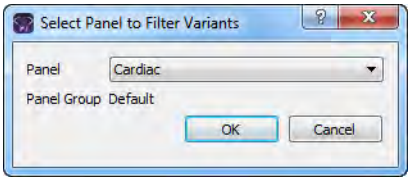
Option	Description
Run Details	Opens the Run tab. See "The Run Tab" on page 139. **Note:** You can also double-click a run entry to open the Run tab.
Add All to Custom Report	Selects all the data that is displayed for the run for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Create Batch Variants Report	Opens the Batch Variants Report dialog box for generating the Batch Variants report for all the variants in all the samples for the selected run. See "To manage a Batch Variants report (Runs tab)" on page 133.
Show Variants of Selected Runs	Displays all the variants in all the samples for the selected run on a single Sample Variant Data sub-tab. CTRL-click to select multiple runs before opening the context menu and selecting this option.
Show Variants of Selected Runs Filtered by Panel	Displays all the variants in all the samples for the selected run filtered by a selected panel in a single Sample Variant Data sub-tab. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected run. CTRL-click to select multiple runs before opening the context menu and selecting this option. *Figure 6-3: Select Panel to Filter Variants dialog box* 
Add Samples to Run	Opens the New Run dialog box so that you can add more samples to the selected run. See "Adding a Run" on page 123.
Delete Run	Delete the selected run from the Geneticist Assistant database.
Export to VCF	Exports all the variants in the selected run to a VCF file. You must specify both the file name and the location in which to save the file.

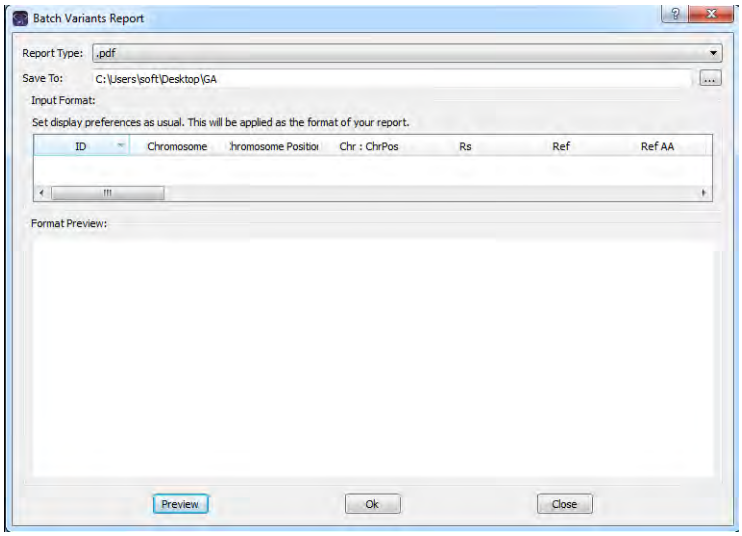
Table 6-1: Runs tab, Runs pane context menu (Continued)

Option	Description
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the run. If you are an Administrator user, then you can set or modify the permissions for a run that any user has submitted. If you are a standard user, then you can set or modify the permissions for only those runs that you have submitted to the Geneticist Assistant database, or for those runs that other users have submitted and to which you have been granted Can Read and Can Write permissions. See "To set or modify the Read/ Write permissions for a run" on page 137.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

To manage a Batch Variants report (Runs tab)

A *Batch Variants report* is a report that includes information for variants over a specified date range or other parameters. A *Batch Variants report configuration* determines the data that is to be included in the report, and if the report is to be available only to the user who created it, or to all users. The configuration does not include the report format (PDF, HTML and so on), or the location in which the report is to be saved. Instead, you must specify the format and location in which to save the report every time you generate a report for a new or saved configuration.

Figure 6-4: Batch Variants Report dialog box



You can generate a Batch Variants report according to a new or saved configuration, modify a Batch Variants report configuration, or delete a Batch Variants report configuration. See:

- "To generate a Batch Variants report based on a new configuration" on page 134.

- “To generate a Batch Variants report based on a saved configuration” below.
- “To modify a Batch Variants report configuration” on page 135.
- “To delete a Batch Variants Report configuration” on page 136.

To generate a Batch Variants report based on a new configuration

When you generate a Batch Variants report, the report name is automatically assigned based on the variant ID for which you are generating the report (for example, MSH2_GC) and you cannot change this. You must select a location in which to save the report.

1. On the Batch Variants Report dialog box, on the Report Type dropdown list, select the report type. (The default value is PDF.)
2. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
   - Leave this location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Optionally, to view an onscreen preview of the report before it is generated and saved, click Preview.
4. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.
5. Click OK.

   The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To generate a Batch Variants report based on a saved configuration

1. On the Batch Variants Report dialog box, select the output format for the report.
2. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
   - Leave the location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.

4. Click Load Configuration.

   The Load Configuration dialog box opens. The dialog box displays all your private and public configurations as well as all the configurations that other users have made public.

Figure 6-5: Load Configuration dialog box

5. Select the configuration that you are loading, and then click Load.

   The Load Configuration dialog box closes. The Select Columns dialog box remains open. The pane display is updated dynamically after you load the configuration.

6. Select a configuration, and then click Load.

   The Load Configuration dialog box closes. The Select Columns dialog box remains open. The dialog box now reflects the columns that are to be included in the report based on the selected configuration.

7. Click Close (x) to close the Select Columns dialog box.

8. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.

9. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.

10. Click OK.

    The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To modify a Batch Variants report configuration

When you modify a Batch Variants report configuration, any reports that were generated according to the “old” (previous) configuration are not affected. Going forward, the report is generated based on the “new” configuration.

1. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank.

2. Do one of the following:
   - Leave this location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.
4. To modify the report configuration, see "To create/modify a pane configuration" on page 32.
5. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.
6. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box opens, with a message that indicates where the report was saved.
7. Click OK.

   The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To delete a Batch Variants Report configuration

You can delete a Batch Variants report configuration only if you created the report. When you delete a Batch Variants report configuration, any reports that were generated according to the "old" (previous) configuration are not affected. Going forward, the "old" configuration is simply not available for selection.

You are not prompted about confirming the deletion of a Batch Variants report configuration, so be very sure that the deletion must be deleted before doing so.

1. On the Batch Variants Report dialog box, right-click on any heading in the Input Format pane.

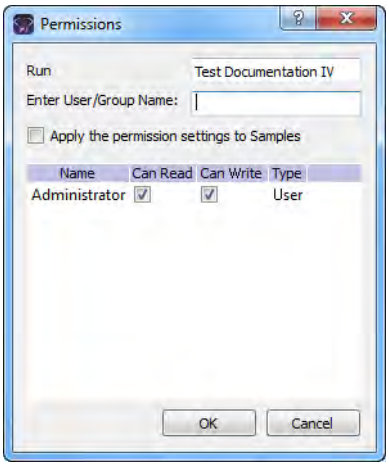
   The Select Columns dialog box opens.
2. See "To delete a pane configuration" on page 34.
3. Click Close (x) to close the Batch Variants Report dialog box.

To set or modify the Read/Write permissions for a run

1. If you have not already done so, right-click the run on the Runs tab, and on the context menu that opens, click Permissions.

   The Permissions dialog box opens. The first time that the dialog box opens, if the run was submitted using a Geneticist Assistant version with a build date of 4/8/15 or later, then the name of the user who submitted the run is displayed in the lower pane of the dialog box and both the Can Read and Can Write permissions are selected. If a standard user submitted the run, then the Administrator user name is also displayed. For runs that were submitted using earlier versions of Geneticist Assistant, the pane is blank. For subsequent openings, the dialog box displays the names of all the users and/or groups and their assigned permissions for the run.

Figure 6-6: Permissions dialog box (Run tab)



2. Modify the Read and Write run permissions for any user or group as needed and then go to Step 6; otherwise, to add run permissions for a new user or group, go to Step 3.

You can add or remove the Can Write permission for a user or group, or you can remove both the Can Read and Can Write permissions. Removing the Can Read permission but leaving the Can Write permission selected for a user or group has no effect.

3. Enter a user name (the login name for the user), or to assign the same permissions to multiple users in a single step, enter a group name.

   As you enter a user name or a group name, a dropdown list opens. The list is dynamically updated with user names or group names that are in your Geneticist Assistant database and that match the search string that you are entering.

4. Select the appropriate user name or group name from the list, and then press [ENTER].

   The user name or group name is displayed in the lower pane of the dialog box. The Read permission is already assigned for the selected user or group. If just this permission is selected, then the user or group can simply view the run. They cannot modify it.

5. Optionally, if the user or group must be able to modify the run as well, then select the Can Write permission.

6. Optionally, if all users and groups are to have their same run permissions applied to *all* the samples in the run, then select Apply the permission settings to Samples; otherwise, leave this option cleared, or clear it if necessary.

You can assign Read and Write permissions to the samples in a run on a per sample basis. See *"To set or modify the Read/Write permissions for the samples in a run on a per sample basis" on page 170.*

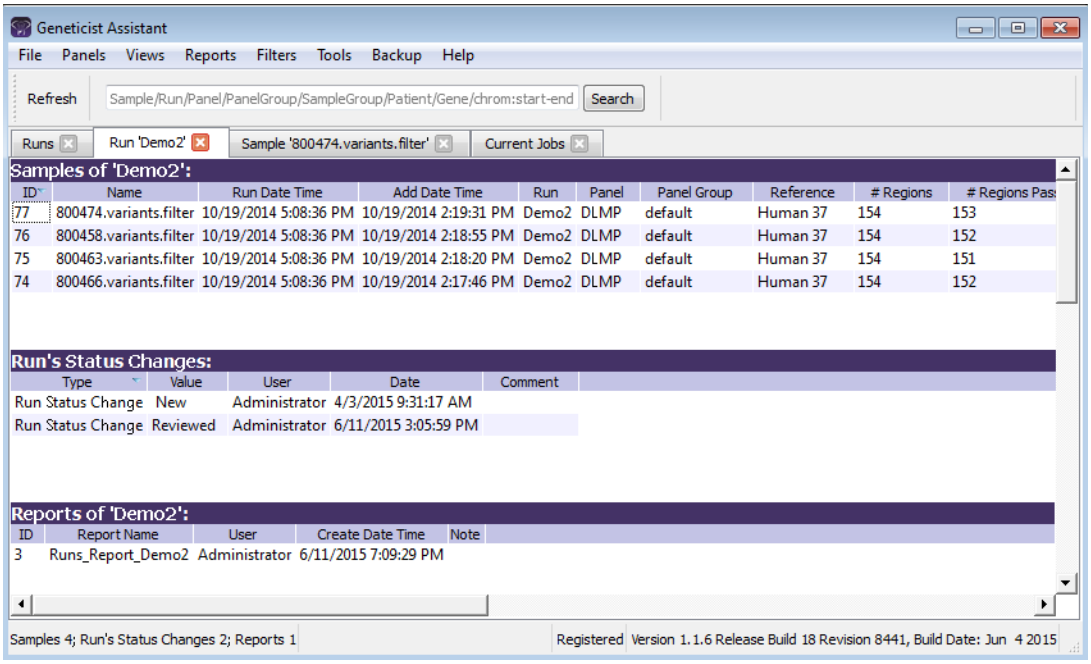
7. Click OK.

The Run Tab

To open the Run tab, do one of the following:

- Right-click a run on the Runs tab, and on the context menu that opens, select Run Details.
- Double-click a run on the Runs tab.

Figure 6-7: Run tab



The Run tab has three panes:

- The Samples pane. See "Samples pane (Run tab)" below.
- The Run's Status Changes pane. See "Run's Status Changes pane" on page 172.
- The Reports pane. See "Reports pane (Run tab)" on page 172.

Samples pane (Run tab)

The Samples pane contains an entry for each sample (VCF/BAM file combination, VCF/DAT file combination, or single VCF file) that was loaded for the run. The following options are available for a sample in the Samples pane on the Run tab:

- Setting the sample status. See "To set the sample status" on page 140.
- Determining missed clinical variants. See "To determine missed clinical variants for a sample" on page 140.
- Using the context menu. See "Samples pane (Run tab) context menu" on page 141.

To set the sample status

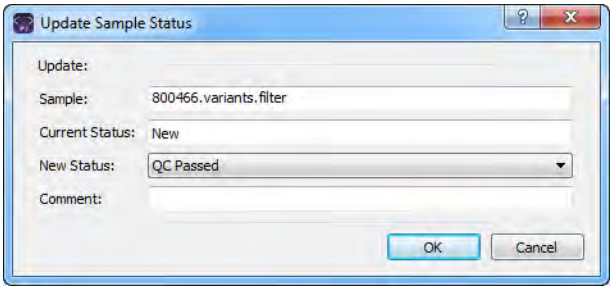
By default, after a sample is initially submitted, the status of the sample is set to New on the Run tab.

1. On the Status dropdown list, select a different status for the sample.

You can select a different value from one of the available default values (New, QC Passed, Reviewed, or Complete), or if needed, you can create custom statuses, and then select one of these custom statuses instead. See "Managing Objects" on page 41.

   The Update Sample Status dialog box opens. The dialog box displays the Current status and the New status, which is the status that you are changing the sample to.

Figure 6-8: Update Sample Status dialog box



2. Optionally, in the Comment field, enter an explanation/reason for changing the sample status.

   The status change and the comment for the status change are displayed in the Status Changes pane on the Sample tab. See "Status Changes pane" on page 185.

3. Click OK.

   The Update Sample Status dialog box closes. The Run tab remains open. The newly selected sample status is displayed in the Status field on the tab.

To determine missed clinical variants for a sample

If any of the clinically relevant positions that are identified in the Clinical Variants (ClinVar) database from the NCBI do not have coverage in the samples, then "Yes" is displayed in the Missed Clinical Variants column in the Samples pane on the Run tab; otherwise, "No" is displayed.

Samples pane (Run tab) context menu

You can right-click a sample in the Samples pane on the Run tab to open a context menu with the following options:

Table 6-2: Run tab, Samples pane context menu

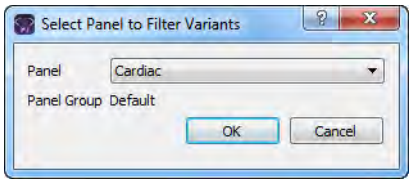
Option	Description
Sample Details	Opens the Sample tab, which contains detailed information about the sample, including the variant list. See "The Sample Tab" on page 173.
Show Sample Variants Filtered by Panel	Displays all the variants in the selected sample filtered by a selected panel. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected run. *Figure 6-9: Select Panel to Filter Variants dialog box* 
Coverage QC	Opens the Coverage QC tab, which displays the coverage information for the sample. See "Coverage QC tab (via the Samples pane on the Run tab)" on page 144.
Coverage Regions	Opens the Coverage Regions tab, which lists the coverage information for every region in your panel. The Status indicates whether the region passed or failed the Quality Control (coverage) settings that were set for the run. **Note:** The information that is shown on this tab is identical to the information that is shown on the Coverage Regions sub-tab on the Coverage QC tab and the context menu for this tab has the same options as those on the context menu for the Coverage Regions sub-tab on the Coverage QC tab. See "Coverage Regions sub-tab (Coverage QC tab)" on page 146. **Note:** The context menu for the tab title bar has the Manage Sample Group Statistics option which you can select if you used sample groups for a run and you want to calculate statistics for the run. This option is also available on the tab title bar context menu for the Sample Variant Data sub-tab on the Sample tab. See "Sample Variant Data sub-tab" on page 173.
Panel Statistics	Opens the Panel Statistics tab. The tab has options for displaying specific information for a selected panel/panel group and a selected sample range. See "Panel Statistics tab (Run tab)" on page 164.

Table 6-2: Run tab, Samples pane context menu (Continued)

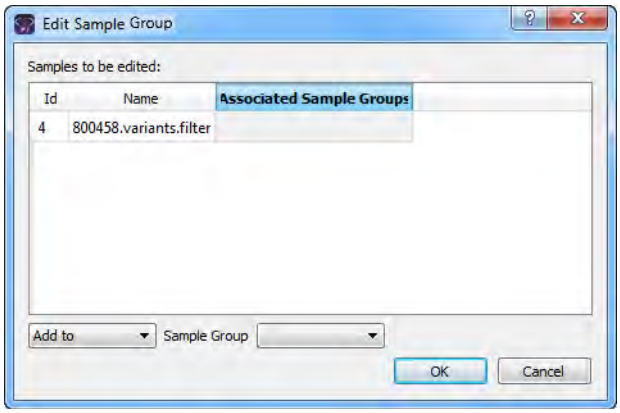
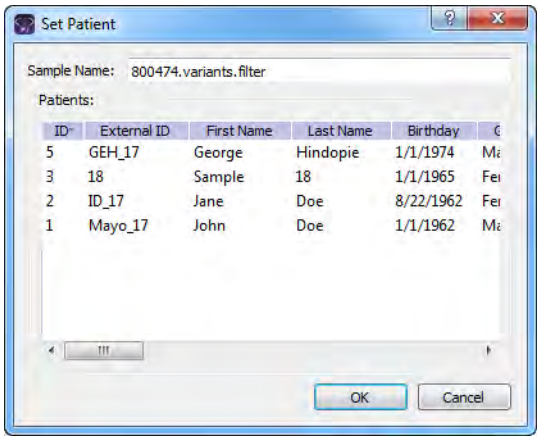
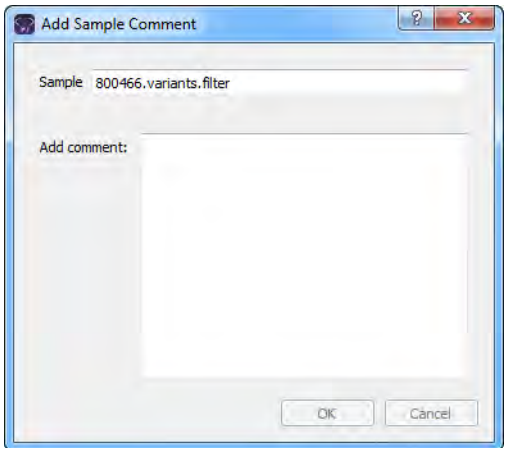
Option	Description
Edit Sample Groups	Opens the Edit Sample Group dialog box. You use the options on this dialog box to add a sample to a selected sample group, or to remove a sample from a selected sample group. **Note:** If the appropriate sample group is not available, you can add it. See "Managing Objects" on page 41. *Figure 6-10: Edit Sample Group dialog box* 
Compare Samples	Displayed only after two or more samples are selected. Opens a Sample Comparison tab that displays coverage, frequency, and read balance information for each sample that is being compared.
Set Patient	Opens the Set Patient dialog box. Select the patient that is to be associated with the sample, and then click OK. *Figure 6-11: Set Patient dialog box*  **Note:** You also have the option of associating a patient with a sample from the Sample tab. See "To associate a patient with a sample" on page 187.
Delete Sample	Deletes the sample from the run.
Add All to Custom Report	Selects all the data that is displayed for all the samples in the run for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.

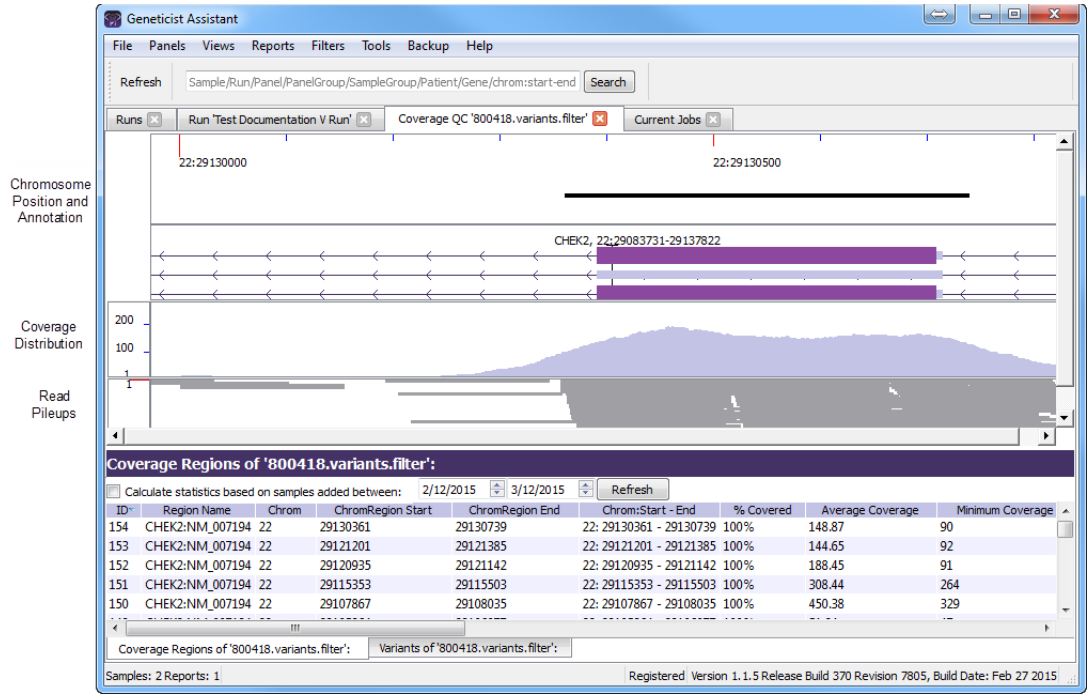
Table 6-2: Run tab, Samples pane context menu (Continued)

Option	Description
Create Batch Variants Report	Opens the Batch Report dialog box for generating the Batch Variants report for all the variants in the selected sample. See "To manage a Batch Variants report (Run tab)" on page 167.
Export to VCF	Exports all the variants in the selected sample to a VCF file. You must specify both the file name and the location in which to save the file.
Add Comment	Opens the Add Sample Comment dialog box. You add a comment for the associated sample in the Add Comment field. The comment is displayed in the Comments field on the Samples sub-tab on the Patient tab, the Comments field in the Samples pane on the Run tab, and in the Comments pane on the Sample Data sub-tab on the Sample tab. *Figure 6-12: Add Sample Comment dialog box*  **Note:** If multiple comments have been added for a sample, then with the exception of the Comments pane on the Sample Data sub-tab on the Sample tab, only the last added comment is displayed in all the locations detailed above. All the comments that have been added for a sample, including the last added, are displayed in reverse chronological order in the Comments pane on the Sample Data sub-tab for the Sample tab.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the sample. If you are an Administrator user, then you can set or modify the permissions for any sample in any run that any user has submitted. If you are a standard user, then you can set or modify the permissions for only those samples that are contained in runs that you have submitted to the Geneticist Assistant database, or for those samples that are contained in runs that were submitted by other users and to which you have been granted Can Read and Can Write permissions. See "To set or modify the Read/Write permissions for the samples in a run on a per sample basis" on page 170.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Coverage QC tab (via the Samples pane on the Run tab)

The Coverage QC tab displays the coverage information for the sample.

Figure 6-13: Coverage QC tab



From top to bottom, the Coverage QC tab contains the following panes:

- Chromosome Position and Annotation pane. See "Chromosome Position and Annotation pane" on page 145.
- Coverage Distribution pane. See "Coverage Distribution pane" on page 145.
- Read Pileups pane. See "Read Pileups pane" on page 145.

The Coverage QC tab also has two sub-tabs that are displayed at the bottom of the tab. See:

- Coverage Regions sub-tab. See "Coverage Regions sub-tab (Coverage QC tab)" on page 146.
- Variants Sub-tab. See "Variants sub-tab (Coverage QC tab)" on page 148.

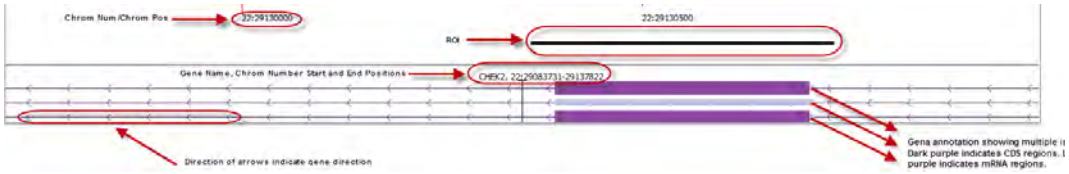
To zoom in or zoom out on a pane display, press and hold the [CTRL] key and use the scroll wheel on the mouse. To move the pane display to the left or right, use the scroll bar at the very bottom of the tab, or click and hold the left mouse button in a pane, and then drag the mouse as needed. All the displays in all the panes are interactive and linked. If you zoom in or zoom out the display in one pane, or move the pane display to the left or the right, then the displays in all the other panes are also updated. When the Coverage QC tab first opens, the first coverage region that is displayed on the Coverage Regions sub-tab is selected and it is the information for this coverage region that is displayed in all the panes. Double-click to select another coverage region on the Coverage Regions sub-tab to change the displays

accordingly, or double-click any variant on the Variants sub-tab to display the region around the variant, with the variant in the center, in the display panes.

Chromosome Position and Annotation pane

The top pane is the Chromosome Position and Annotation pane. The top half of this pane shows the Chromosome Region Start and End positions for the region that is currently selected on the Coverage Regions sub-tab. The black horizontal bars show the panel region within this displayed region. The gene annotation for the chromosome region is displayed below the chromosome information in the bottom half of the pane. The top line of the gene annotation shows the gene name, and the chromosome number:chromosome position for the gene. Each horizontal line below the gene name indicates a different isoform of the gene, with the arrows on the horizontal line indicating the gene direction. An arrow that points to left (<-) indicates a gene that is found on the negative (-) strand. An arrow that points to the right (->) indicates a gene that is found on the positive (+) strand. The dark purple shading indicates a CDS region and the light purple shading indicates an mRNA region.

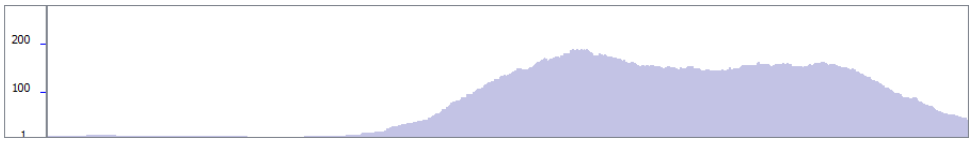
Figure 6-14: Coverage QC tab, Chromosome Position and Annotation pane



Coverage Distribution pane

The middle pane is the Coverage Distribution pane. The Coverage Distribution pane displays coverage information for the region that is currently selected on the Coverage Regions sub-tab. The height of the region indicates the coverage depth.

Figure 6-15: Coverage QC tab, Coverage Distribution pane



Read Pileups pane

The bottom pane is the Read Pileups pane. The Read Pileups pane displays all the reads that are aligned to the region that is currently selected on the Coverage Regions sub-tab.

Figure 6-16: Coverage QC tab, Read Pileups pane

Coverage Regions sub-tab (Coverage QC tab)

The Coverage Regions sub-tab has a single pane, the Coverage Regions pane, that displays the coverage information for every region in your panel. The Status column indicates whether the panel region passed or failed the Quality Control (coverage) settings that were set for the run's panel. By default, the sub-tab displays the coverage statistics for the samples that were added within the last 30 days based on the current day's date. You can change the date, and then click Refresh to calculate the statistics based on this new date range.

Figure 6-17: Coverage Regions sub-tab

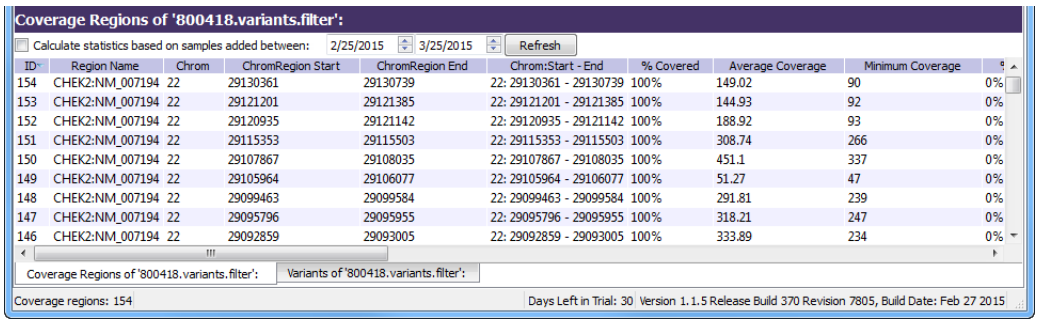


Table 6-3: Coverage QC tab, Coverage Regions sub-tab

Column	Description
Region Name	The region description from Column 4 of the BED file.
Chrom	The chromosome on which the region is located.
ChromRegionStart	The starting chromosome position for the region.
ChromRegionEnd	The ending chromosome position for the region.
Note: Together, the ChromRegionStart and ChromregionEnd make up the chromosome position range.	
Chrom:Start-End	The chromosome position range appended to the chromosome number.
% Covered	The percentage of the region that has reads aligned to it.
Average Coverage	The average coverage (average number of aligned reads) for the region.
Minimum Coverage	The minimum coverage (number of reads aligned) for any position within the region.
% Covered Threshold	The percentage of the region that must have reads aligned to it, or the region does not pass the Quality Control settings that have been set for the run's panel. See Chapter 4, "Managing Panels," on page 77.
Average Coverage Threshold	The average coverage (average number of aligned reads) required for the region, or the region does not pass the Quality Control settings that have been set for the run's panel. See Chapter 4, "Managing Panels," on page 77..
Minimum Coverage Threshold	The minimum coverage (number of reads aligned) that required for any position within the region, or the region does not pass the Quality Control settings that have been set for the run's panel. See Chapter 4, "Managing Panels," on page 77.

Table 6-3: Coverage QC tab, Coverage Regions sub-tab (Continued)

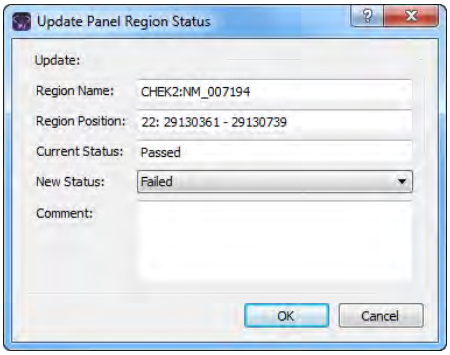
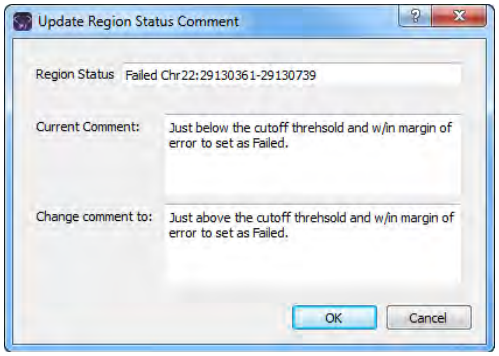
Column	Description
Status	Indicates if the region passed or failed the Quality Control (coverage) settings that were specified for the run's panel. You can manually change the status for a region. If you manually change the status for a region, then the Update Panel Region dialog box opens. The dialog box lists the new status for the region, and you have the option of adding comments for the status change. The comments are displayed in the Status Comment field. *Figure 6-18: Update Panel Region Status dialog box*  **Note:** By default, only two statuses, Passed or Failed, are available, and one of these two statuses is always automatically selected for the region. You have the option of adding custom Panel Region statuses and manually selecting a status for a region. See "Managing Objects" on page 41.
Status Comment	Displays any comments that were added when the region status is manually changed. You can double-click in this field to add a new comment or modify an existing comment. After you add or edit a comment, and then click out of the field, the Update Region Status Comment dialog box opens. *Figure 6-19: Update Status Region Comment dialog box*  The dialog box displays the status and region positions for the selected region, the current (old) comment, and the new comment. You can make any additional edits to the new comment as needed, and then click OK to close the dialog box and add the new or modified comment for the region.
Average % Covered	Average of the percent covered for the region across all samples for the panel.
Average Average Coverage	The average of the average coverage values for the region across all samples for the panel.

Table 6-3: Coverage QC tab, Coverage Regions sub-tab (Continued)

Column	Description
Average Minimum Coverage	The average minimum coverage for the region across all samples for the panel.
Passed	The total number of samples for the panel for which the status of the region is Passed.
Passed Percent	The percent of the samples for the panel for which the status of the region is Passed.
Failed	The total number of samples for the panel for which the status of the region is Failed.
Total	The total number of samples that were submitted for the panel.
Gene	The gene on which the region is located.
Transcript	The accession number for the mRNA transcript.
Protein	The accession number for the protein transcript.

You can right-click anywhere on the Coverage Regions sub-tab to open a context menu with the following options:

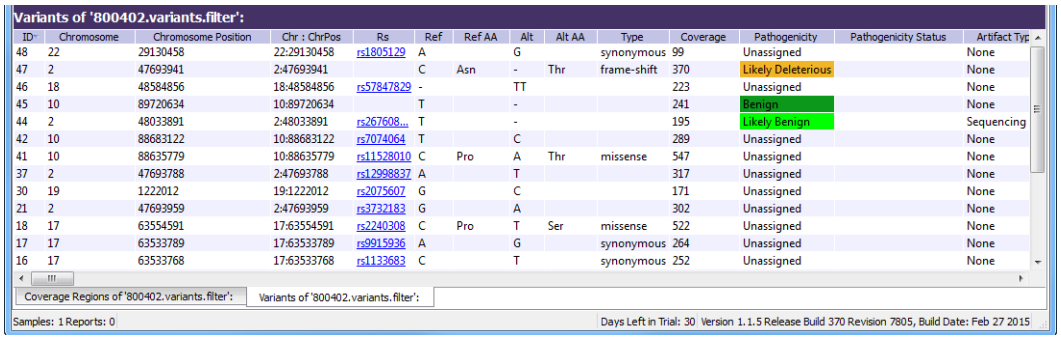
Table 6-4: Coverage QC tab, Coverage Regions sub-tab context menu

Option	Description
Add Selected Regions to Action List	Adds the selected panel region to an Action List. Press and hold the CTRL key to select multiple regions. See "To add selected panel regions or variants to an Action list" on page 162.
Add Selected Regions to Custom Report	Selects all the data that is displayed for the selected panel region for inclusion in a custom report. Press and hold the CTRL key to select multiple regions. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Add All to Custom Report	Selects all the data that is displayed for all the panel regions for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the sub-tab. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Variants sub-tab (Coverage QC tab)

The Variants sub-tab has a single pane, the Variants pane, that displays all the variants that were detected in the sample. (See Figure 6-20 on page 149.) By default, two types of information are displayed on the Variants sub-tab—variant annotation information and global variant count and frequency information. Table 6-5 on page 149 details *all* the variant annotation information that is shown by default for each variant. Table 6-6 on page 154 details all the global variant count and frequency information that is shown by default for each variant. You can double-click on any variant on the Variants sub-tab to display the region around the variant, with the variant in the center in the display panes.

Figure 6-20: Coverage QC tab, Variants sub-tab



The information that is displayed on the Variants sub-tab was divided into two tables simply for ease of reading in this manual and locating and identifying the information on the sub-tab. There is no visible division of this information on the sub-tab itself.

- *Remember, you can select which columns to show and hide in a pane. See "Work with columns in a tab pane" on page 30.*
- *Remember, you can specify how the information about the variants is displayed in the Variants Table sub-tab. See "To specify variant preferences" on page 53.*
- *Selected columns from the various reference information panes on the Variant tab can also be displayed here. See "The Variant Tab" on page 190.*

Table 6-5: Coverage QC tab, Variants sub-tab: Variant Annotation information

Column	Description
ID	System-generated value that identifies the variant.
Chromosome	The chromosome on which the variant is located.
Chromosome Position	The chromosome position where the variant is located.
Chr:ChrPos	The chromsome:chromosome position for the variant location.
Rs	The dbSNP database identification for the variant. **Note:** If a value other than N/A is displayed, then it is a hyperlink to the NCBI dbSNP web page for the variant.
Ref	The reference nucleotide at the variant position.
Ref AA	The reference amino acid coded for by the codon that includes the variant position.
Alt	The variant nucleotide at the variant position.
Alt AA	The variant amino acid coded for by the codon that includes the variant position.
Type	The type of variant based on the amino acid change that the variant caused. Values are Missense, Synonymous, Frameshift, or Nonsense.
Coverage	The coverage (number of reads) aligned to the variant location in the sample.

Table 6-5: Coverage QC tab, Variants sub-tab: Variant Annotation information (Continued)

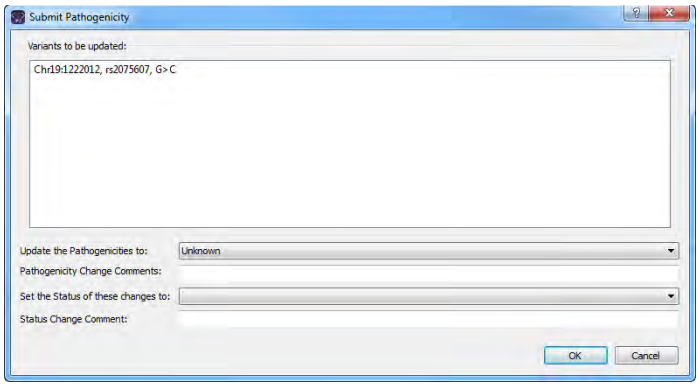
Column	Description
Pathogenicity	The pathogenicity of the variant, which is the potential for the variant to cause disease. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be; for example, Deleterious. (If you cannot determine the pathogenicity of the variant even after reviewing all the relevant information, you can set the status to Unknown.) After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant. To open the Submit Pathogenicity Status dialog box, double-click in this column for the selected variant. You can update the pathogenicity for the selected variant and/or the status of the pathogenicity in this dialog box. You can also add a comment about the pathogenicity change and/or pathogenicity status change **Note:** The option to update the pathogenicity and/or pathogenicity status ("Update Pathogenicity") is also available on the context menu for the pane. *Figure 6-21: Submit Pathogenicity dialog box*  The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See "Managing Objects" on page 41. • Optionally, you can add supporting information for this pathogenicity change. See "To add supporting information for a pathogenicity change" on page 198. • You always have the option of updating just the pathogenicity status for a variant at a later date. See "Pathogenicity Status" below.

Table 6-5: Coverage QC tab, Variants sub-tab: Variant Annotation information (Continued)

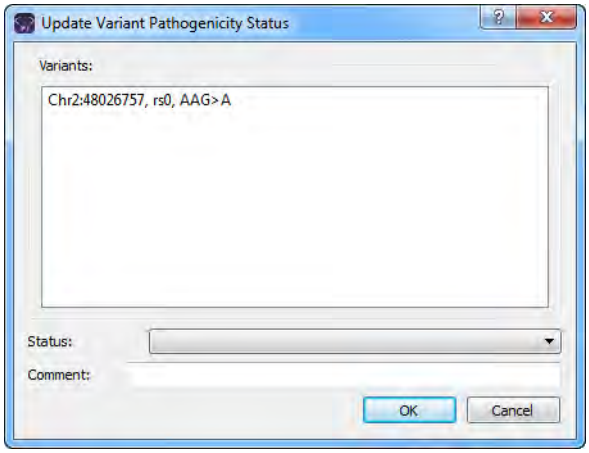
Column	Description
Pathogenicity Status	The status of the selected pathogenicity. You must manually set this value.To open the Update Variant Pathogenicity Status dialog box, double-click in this column for the selected variant. You can set just the pathogenicity status for the variant in this dialog box. You can also add a comment about the status change. **Note:** The option to update just the status (“Update Pathogenicity Status”) is also available on the context menu for the pane. *Figure 6-22: Update Variant Pathogenicity Status dialog box* 

Table 6-5: Coverage QC tab, Variants sub-tab: Variant Annotation information (Continued)

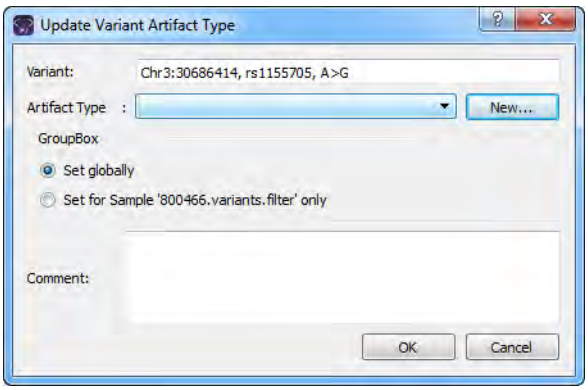
Column	Description
Artifact Type	The artifact type for the selected variant. You must manually set this value. Double-click in this column for the selected variant to open the Update Artifact Type dialog box. You specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type. **Note:** The option to update the artifact type (“Update Artifact Type”) is also available on the context menu for the pane. *Figure 6-23: Update Variant Artifact Type dialog box*  When you specify the artifact type, you can set the artifact type as the following: • Set globally—Selected by default. Set this artifact type for the variant, regardless of the sample in which the variant is detected. • Set for Sample—Set this artifact type for this variant only for the indicated sample. You also have the option of adding comments for the variant artifact type. The following information is relevant to updating the artifact type for a variant: • You can press and hold the CTRL key to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant on the Variants sub-tab on the Patient tab, on the Variants sub-tab on the Panel Statistics tab via the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See “Managing Objects” on page 41.
Variant Frequency	The frequency at which the variant was detected in the sample. Any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.
Zygosity	Indicates whether the variant is heterozygous or homozygous. **Note:** The selected zygosity is based on the Zygosity settings that you have specified on the Variant Preferences tab of the Settings dialog box. See “To specify variant preferences” on page 53.
Read Balance	The balance in forward and reverse oriented reads with the variant as reported in the VCF file.
Gene	The gene on which the variant is located.
Gene Strand	The strand (+ or -) for the gene in which the variant is located.
Exon Number	The exon within the gene in which the variant is located. “Intron” is displayed for intronic variants.

Table 6-5: Coverage QC tab, Variants sub-tab: Variant Annotation information (Continued)

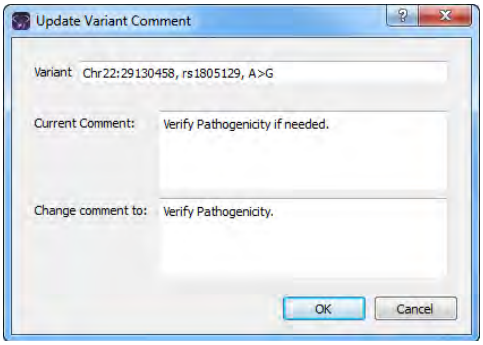
Column	Description
Transcript	The accession number for the mRNA transcript that was used for variant calling.
Protein	The accession number for the protein transcript that was used for variant calling.
Coding Base	The position in the coding regions for the gene where the variant is located.
Codon Position	The position within the codon where the variant is located (1, 2, or 3).
AA Position	The amino acid number within the gene for the amino acid coded for by the codon that includes the variant position.
HGVS Genomic	The HGVS genomic nomenclature for the variant relative to the reference accession that is shown in the Genomic column. (www.hgvs.org/mutnomen).
HGVS Coding	The HGVS coding DNA nomenclature for the variant relative to the reference accession that is shown in the Transcript column. (www.hgvs.org/mutnomen).
HGVS Protein	The HGVS protein nomenclature for the variant relative to the reference accession that is shown in the Protein column. (www.hgvs.org/mutnomen).
Variant Comment	The last added comment for the variant. You can also double-click in this field, and then add or edit a comment as needed. After you click out of the field, the Update Variant Comment dialog box opens. *Figure 6-24: Update Variant Comment dialog box*  The dialog box displays the variant for which you are adding or editing the comment, the current (old) comment, and the new comment. You can make additional edits to the new comment needed, and then click OK to close the dialog box and save the new or modified comment for the variant. **Note:** You can also add comments for a variant with an option on the context menu for this sub-tab (see "Add Comment" below), **Note:** If multiple comments have been added for a variant, then with the exception of the Variant Comments pane on the Variant tab, only the last added comment is displayed here and on the Variants sub-tab on the Patient tab, the Sample Variant Data sub-tab for the Sample tab, the Variant pane on the Variant tab, and the Variant Comments pane on the Variant tab. All the comments that have been added for a variant, including the last added, are displayed in reverse chronological order in the Variant Comments pane on the Variant tab.
Times Observed Per Run	The number of times that the variant was detected across all samples in the run.
Panel	The panel used for the run.
Panel Group	The panel group to which the panel is assigned.

Table 6-5: Coverage QC tab, Variants sub-tab: Variant Annotation information (Continued)

Column	Description
Trans	Indicates whether the variant is Transition (Ti) or Transversion (Tv).
Sample	The sample file name.
HGMD Mutation	The amino acid changes that was caused by the variant as recorded in HGMD.
HGMD HGVS	The HGVS code relative to HGMD.
HGMD ID	The ID for the variant in HGMD.
Note: If you have not specified your login settings for the HGMD database (see "To specify HGMD settings" on page 56), then these last three columns are not displayed.	

Table 6-6: Coverage QC tab, Variants sub-tab: Global Variant Count and Frequency information

Column	Description
Samples Per Panel	The total number of samples submitted per panel.
Coverage Region Status	Indicates whether the region passed or failed the Quality Control (coverage) settings that were specified for the run's panel.
Note: Any of the following values that can also be reported as real or related to the artifact type for the variant are marked with an asterisk. For example, for Times Observed Per Panel, two additional columns are also displayed—Times Observed As Real Per Panel and Times Observed As Artifact Per Panel, where the number of samples for which the Artifact Type is set to None is reported for any Observed as Real value and the number of samples for which the Artifact Type is set to any other value, such as Sequencing, is reported for any Observed as Artifact value.	
Times Observed Per Panel*	The number of times that the variant was detected across all samples per panel.
Frequency Observed Per Panel*	The frequency at which the variant was detected across all samples per panel.
Samples Per Patient Per Panel	The number of samples that are included in the Geneticist Assistant database for the patient who is associated with the selected sample relative to the panel that is assigned for the selected sample.
Times Observed Per Patient Per Panel*	The number of times that the variant was detected across all samples per panel group.
Frequency Observed Per Patient Per Panel*	The frequency at which the variant was detected for the patient who is associated with the selected sample relative to the panel that assigned for the selected sample.
Samples Per Panel Group	The number of samples that are included in the panel group that include the panel that is assigned to the selected sample.
Times Observed Per Panel Group*	The number of samples that include the variant and that are assigned to any panel that is included in the same panel group as the panel for the selected sample.
Frequency Observed Per Panel Group*	The frequency at which the variant was detected in all samples that are assigned to any panel that is included in the same panel group as the panel for the selected sample.
Patient Variant Frequency	The frequency of the variant found in the patient sample. If multiple samples for a patient have been loaded, then all samples are considered.
Samples With Passed Regions Per Panel	The number of samples that are assigned to the panel for the selected sample where the panel region in which the variant is located has a status of Passed for the sample.

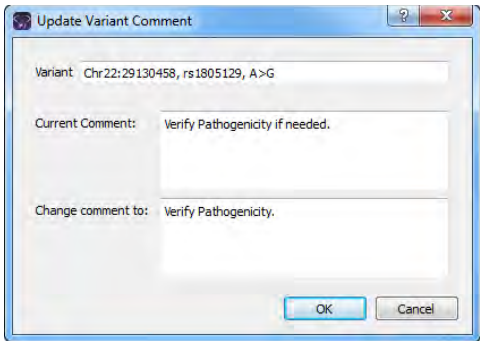
Table 6-6: Coverage QC tab, Variants sub-tab: Global Variant Count and Frequency information (Continued)

Column	Description
Times Observed In Passed Regions Per Panel	The number of samples that are assigned to the panel for the selected sample where the variant is reported and the panel region in which the variant is located has a status of Passed for the sample.
Frequency Observed In Passed Regions Per Panel	The frequency at which the variant was detected considering only those samples that are assigned to the same panel as the selected sample and where the panel region in which the variant is located has a status of Passed.
Samples With Passed Regions Per Patient Per Panel	The number of samples that are assigned to the patient and panel that are assigned to the current sample where the panel region in which the variant is located has a status of Passed.
Times Observed In Passed Regions Per Patient Per Panel	The number of samples that are assigned to the patient and panel that are assigned to the current sample where the variant is found and the panel region in which the variant is located has a status of Passed.
Frequency Observed In Passed Regions Per Patient Per Panel	The frequency at which the variant was detected in all samples that are assigned to the patient and panel that are assigned to the current sample, and the panel region has a status of Passed.
Samples With Passed Regions Per Panel Group	The number of samples that are assigned to any panel that is included in the same panel group as the panel for the selected sample where the panel region in which the variant is located has a status of Passed.
Times Observed In Passed Regions Per Panel Group	The number of samples that are assigned to any panel that is included in the same panel group as the panel for the selected sample where the variant is found and the panel region in which the variant is located has a status of Passed.
Frequency Observed In Passed Regions Per Panel Group	The frequency at which the variant was detected in all samples that are assigned to any panel that is included in the same panel group as the panel for the selected sample where the panel region in which the variant is located has a status of Passed.

You can do the following for a variant that is displayed on the sub-tab:

- You can specify how the information about the variants is to displayed on this sub-tab. See "To specify variant preferences" on page 53.
- You can select a different transcript for the variant on the Transcript dropdown list.
- You can single click twice in the Variant Comment field to select the current comment in the field and edit it, or if the field is blank, enter a new comment. After you add or edit a comment, and then click out of the field, the Update Variant Comment dialog box opens. See Figure 6-25 on page 156.

Figure 6-25: Update Variant Comment dialog box



The dialog box displays the variant for which you are adding or editing the comment, the current (old) comment, and the new comment. You can make additional edits to the new comment needed, and then click OK to close the dialog box and save the new or modified comment for the variant.

You can also use the Add Variant Comment option that is available on the context menu for the variant to add a new variant comment. See "Add Comment" on page 158.

- You can click once in the Type field, the Zygosity field, the HGVS Genomic field, the HGVS Coding field, the HGVS Protein field, and/or the Read Balance field to select the current value in the field and edit it, or if the field is blank, enter a new value.

If you make a change to any of these values for the variant, then these changes are recorded for the variant on the Variant Changes pane on the Variant tab. See "Variant Changes pane" on page 196. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in the Geneticist Assistant application.

- You can right-click a variant in the pane to open a context menu with the following options:

Table 6-7: Coverage QC tab, Variants sub-tab context menu

Option	Description
View in Alamut	Opens the variant for viewing in Alamut. **Note:** To view a variant in Alamut, your Alamut settings must be specified. See "To specify Alamut settings" on page 54.

Table 6-7: Coverage QC tab, Variants sub-tab context menu (Continued)

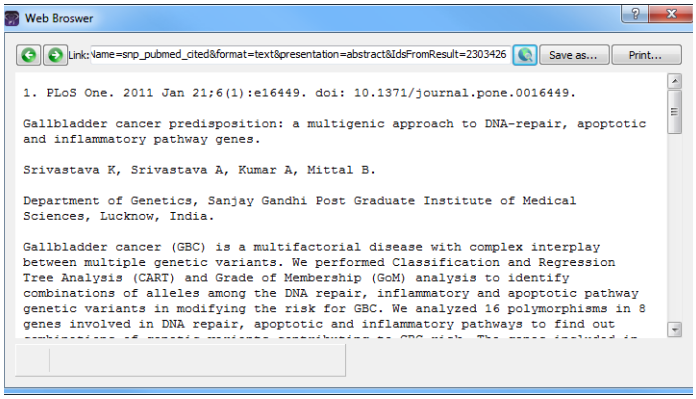
Option	Description
View PubMed Abstract	Searches PubMed for the variant and displays the appropriate abstract in the PubMed window. *Figure 6-26: PubMed window*  You can do the following for the abstract: • In the Link field, enter a different PubMed URL, and then click the Go To icon to navigate to abstract. • Click Save As to save the abstract as a PDF with a name and in a location of your choosing. Click Print to print the abstract.
Add Selected Variants to Action List	Adds the selected variant to an Action List. Press and hold the CTRL key to select multiple variants. See “To add selected panel regions or variants to an Action list” on page 162.
Add Selected Variants to Custom Report	Selects all the data that is displayed for the selected variant for inclusion in a custom report. Press and hold the CTRL key to select multiple variants. See Chapter 7, “Geneticist Assistant Reports,” on page 203.
Add All to Custom Report	Selects all the data that is displayed for all the variants for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.

Table 6-7: Coverage QC tab, Variants sub-tab context menu (Continued)

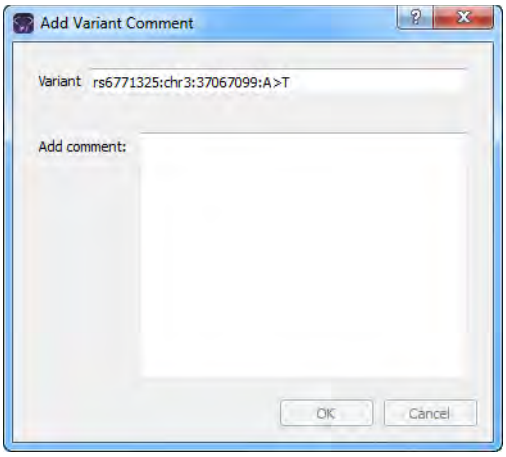
Option	Description
Add Comment	Opens the Add Variant Comment dialog box. You can add a comment for the variant in the Add Comment field, and then click OK. The comment is displayed in the Variant Comment field in the Variants pane on the Search Results tab, the Variant Comment field on the Variants sub-tab on the Patient tab, the Variant Comment field on the Variants sub-tab on the Coverage QC tab, the Variant Comment field on the Sample Variant Data sub-tab on the Sample tab, the Variant Comment field in the Variant pane on the Variant tab, and the Variant Comments pane on the Variant tab. *Figure 6-27: Add Variant Comment dialog box*  **Note:** If multiple comments have been added for a variant, then with the exception of the Variant Comments pane on the Variant tab, only the last added comment is displayed in all the locations that are detailed above. All the comments that have been added for a variant, including the last added, are displayed in reverse chronological order in the Variant Comments pane on the Variant tab.

Table 6-7: Coverage QC tab, Variants sub-tab context menu (Continued)

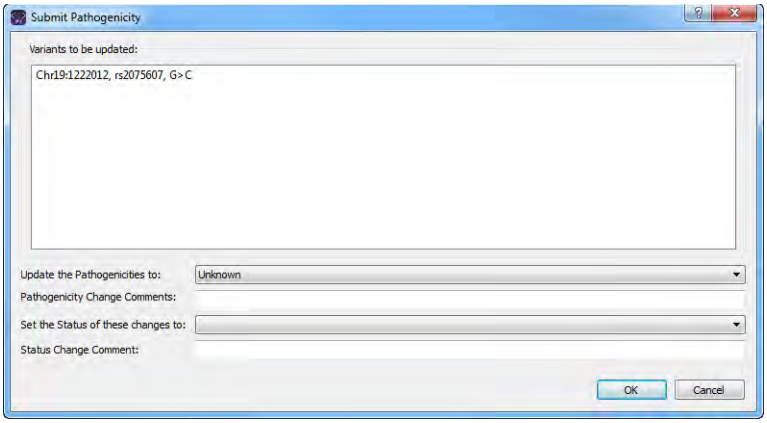
Option	Description
Update Pathogenicity	Opens the Submit Pathogenicity dialog box. You can update the pathogenicity for the selected variant and/or the status of the pathogenicity in this dialog box. You can also add a comment about the pathogenicity change and/or pathogenicity status change. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be; for example, Deleterious. (If you cannot determine the pathogenicity of the variant even after reviewing all the relevant information, you can set the status to Unknown.) You must review the information for the variant and then decide what the pathogenicity should be. After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant. *Figure 6-28: Submit Pathogenicity dialog box*  **Note:** You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box.You can also press and hold the CTRL key to select multiple variants for updating the pathogenicities and the pathogenicity statuses at the same time. **Note:** You can also update the Pathogenicity and the Pathogenicity Status for a variant on the Variants sub-tab on the Patient tab and the Sample Variant Data sub-tab on the Sample tab. The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See “Managing Objects” on page 41. • Optionally, you can add supporting information for this pathogenicity change. See “To add supporting information for a pathogenicity change” on page 198. • You always have the option of updating just the pathogenicity status for a variant at a later date. See “Update Pathogenicity Status” below.

Table 6-7: Coverage QC tab, Variants sub-tab context menu (Continued)

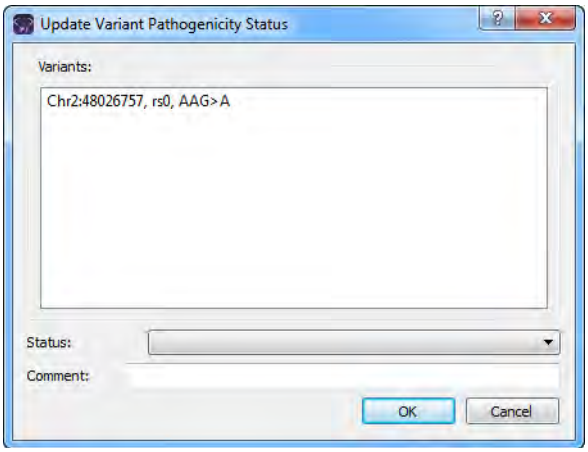
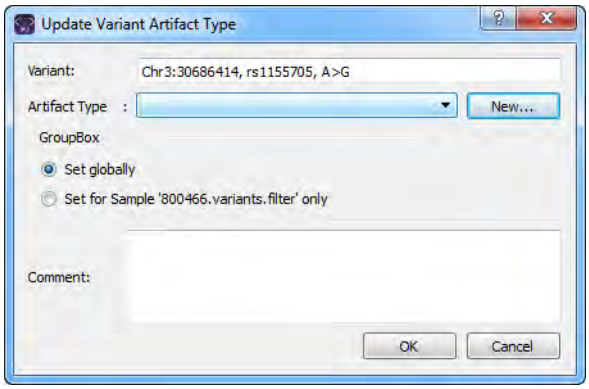
Option	Description
Update Pathogenicity Status	Opens the Update Variant Pathogenicity Status dialog box. You can update the pathogenicity status for the variant in this dialog box. You can also add a comment for the variant. *Figure 6-29: Update Variant Pathogenicity Status dialog box*  **Note:** By default, only two statuses, Pending or Confirmed, are available. You have the option of adding custom pathogenicity statuses. See "Managing Objects" on page 41. **Note:** You can also update the Pathogenicity Status for a variant on the Variants sub-tab on the Patient tab and the Sample Variant Data sub-tab on the Sample tab. **Note:** You can also double-click in the Pathogenicity Status column for a variant to open the Update Variant Pathogenicity Status dialog box. You can also press and hold the CTRL key to select multiple variants for updating the pathogenicity statuses at the same time.

Table 6-7: Coverage QC tab, Variants sub-tab context menu (Continued)

Option	Description
Update Artifact Type	Opens the Update Artifact Type dialog box. You can specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type. *Figure 6-30: Update Variant Artifact Type dialog box*  When you specify the artifact type, you can set the artifact type as the following: • Set globally—Selected by default. Set this artifact type for the variant, regardless of the sample in which the variant is detected. • Set for Sample—Set this artifact type for this variant only for the indicated sample. You also have the option of adding comments for the variant artifact type. The following information is relevant to updating the artifact type for a variant: • You can press and hold the CTRL key to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant on the Variants sub-tab on the Patient tab, on the Variants sub-tab on the Panel Statistics tab via the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See "Managing Objects" on page 41.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the sub-tab. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations. **Note:** If you set column display preferences here, then you can apply these same display preferences for variants in all the variant sub-tabs and panes (with the exception of the Sample Group columns on the Sample Variant Data sub-tab on the Sample tab and the Variant pane on the Variant tab) in a single step. See "To apply shared preferences or to clear all tab preferences" on page 51.

To add selected panel regions or variants to an Action list

An *Action list* is a list of items that require some type of follow up. You can create multiple action lists relative to the type of follow up that is needed. You can create an Action List for panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab or for variants on the Variants sub-tab on the Coverage QC tab or the Sample Variant Data sub-tab on the Sample tab. For example, if several panel regions failed the coverage settings for a run sample, then you can make an Action List that consists of these regions and indicate that you want to review the coverage values for these regions.

1. Do one of the following:

   - To select a single panel region for the Action list, right-click the appropriate panel region on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab, and on the context menu that opens, click Add Selected Regions to Action List.
   - To select multiple panel regions, press and hold the CTRL key as you select the appropriate panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab, and then right-click any selected panel region, and on the context menu that opens, click Add Selected Regions to Action List.
   - To select a single variant for the Action list, right-click on the appropriate variant on the Variants sub-tab on the Coverage QC tab or the Sample Variant Data sub-tab on the Sample tab, and on the context menu that opens, click Add Selected Variants to Action List.
   - To select multiple variants, press and hold the CTRL key as you select the appropriate variants on the Variants sub-tab on the Coverage QC tab or the Sample Variant Data sub-tab on the Sample tab, and then right-click on any selected variant, and on the context menu that opens, click Add Selected Variants to Action List.

   The Add To Action List dialog box opens. If the Action Type that you want to use is already available on the Action type dropdown list, then go to Step 5; otherwise, if this is the first time that you are creating an Action List for the pane or the Action Type that you want to use is not available on the Action Type dropdown list, then you must add at least one type to the list. Go to Step 2.

Figure 6-31: Add To Action List dialog box

2. Click New Type.

   The Add To Action List dialog box is refreshed with an option for adding an action type.

3. In the New Type field, enter an action type (for example, Review Coverage), and then click Add.

   The action type is added to the Action Type list. The Add to Action List dialog box is refreshed to show the full Action Type dropdown list and Comment field.

4. Repeat Step 2 and Step 3 as needed to add more action types.

5. On the Action Type dropdown list, select the appropriate action type.

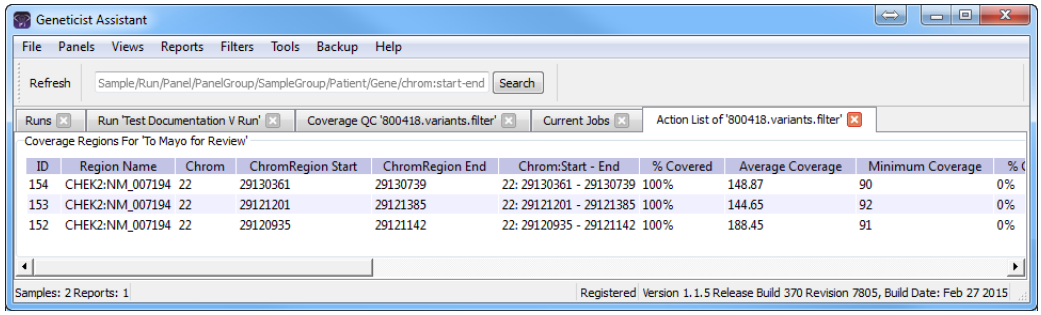
If you are creating an Action List for panel regions, then only the action types that were created specifically for panel regions are available. Likewise, if are creating an Action List for variants, then only the action types that were created specifically for variants are available.

6. Optionally, in the Comment field, enter a comment that explains/supports your decision for adding the selected panel regions or variants to an Action list.

7. Click OK.

   The Add To Action List dialog box closes. The Action List tab for the sample opens. Different panes (grouped by Action Type) list any panel regions for the sample that have been selected for an Action list and any variants for the sample that have been selected for an Action list.

Figure 6-32: Action List tab



To save an Action List, you can generate a standard Action List Report. If multiple panes are included on the Action List tab, then by default, all these panes are included in the report. To remove a pane from the report, before generating the report, right-click on the pane that you want to remove, and then click Remove. See "Generating a Standard Report" on page 205.

Panel Statistics tab (Run tab)

The Panel Statistics tab has options for displaying specific information for a selected panel/ panel group and a selected sample range for samples that have been added within the last 30 days of the current day's date, or a custom date range.

After you change any of the display options for the Panel Statistics tab, you must click Refresh to update the display accordingly.

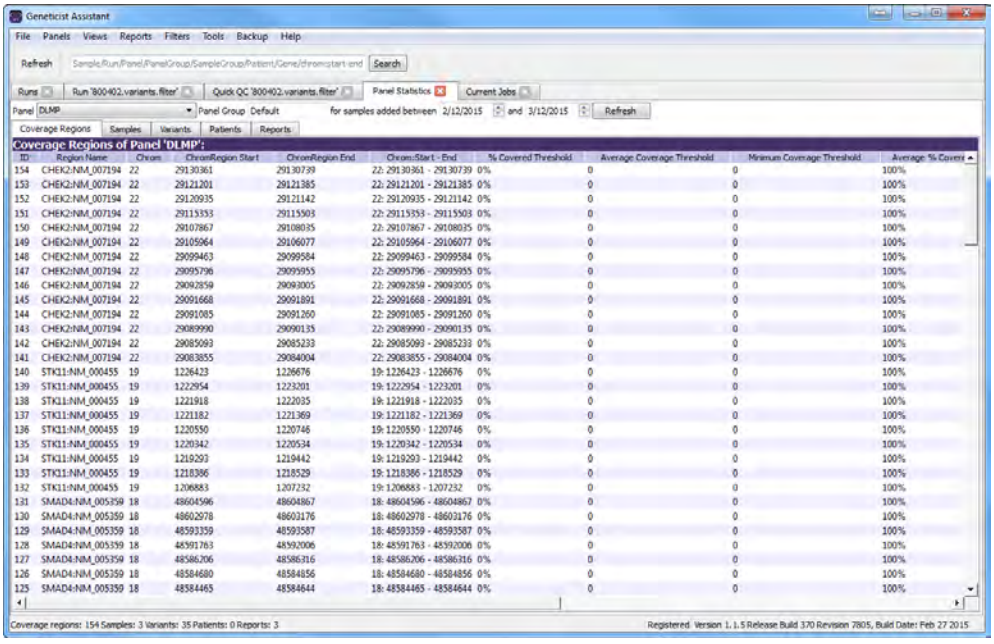
Five sub-tabs are available for the Panel Statistics tab:

- Coverage Regions sub-tab. See "Coverage Regions sub-tab (Panel Statistics tab via the Run tab)" below.
- Samples sub-tab. See "Samples sub-tab (Panel Statistics tab via the Run tab)" on page 165.
- Variants sub-tab. See "Variants sub-tab (Panel Statistics tab via the Run tab)" on page 165.
- Patients sub-tab. See "Patients sub-tab (Panel Statistics tab via the Run tab)" on page 166.
- Reports sub-tab. See "Reports sub-tab (Panel Statistics tab via the Run tab)" on page 166.

Coverage Regions sub-tab (Panel Statistics tab via the Run tab)

The Coverage Regions sub-tab has a single pane, the Coverage Regions pane, that lists the coverage information across all samples for every region in the selected panel/panel group and for the indicated date range. See "Coverage Regions sub-tab (Coverage QC tab)" on page 146 for a detailed discussion of the information that is displayed on this sub-tab.

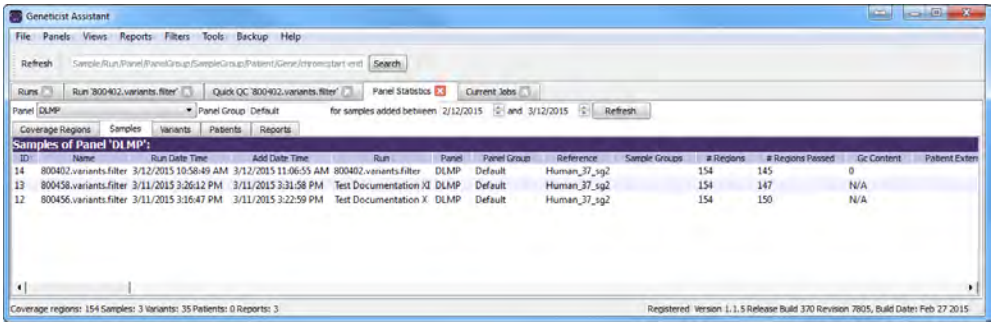
Figure 6-33: Coverage Regions sub-tab (Panel Statistics tab)



Samples sub-tab (Panel Statistics tab via the Run tab)

The Samples sub-tab has a single pane, the Samples pane, that lists all the samples that were imported within the indicated date range for the selected panel/panel group. You can double-click a sample on this sub-tab to open the Sample tab. (See “The Sample Tab” on page 173.) You can also right-click a sample on the sub-tab to open a context menu that has options that are identical to options that are on the context menu for the Samples pane on the Run tab. See “Samples pane (Run tab) context menu” on page 141.

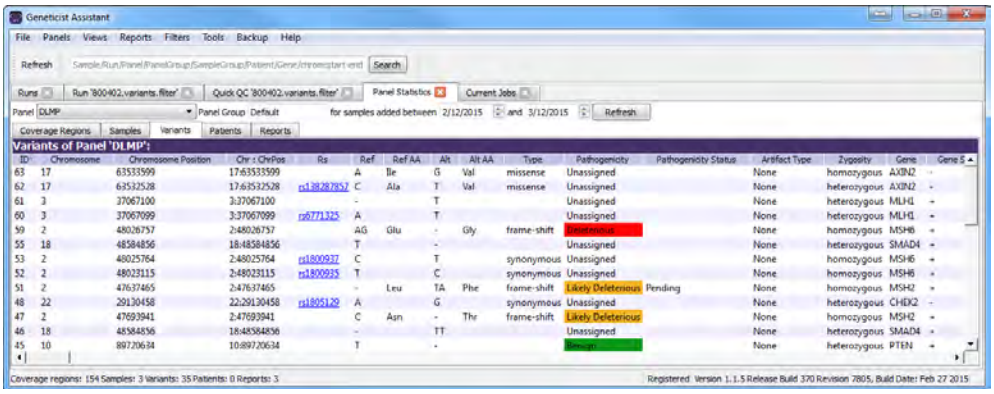
Figure 6-34: Panel Statistics tab, Samples pane



Variants sub-tab (Panel Statistics tab via the Run tab)

The Variants sub-tab has a single pane, the Variants pane, that lists the all variants that were detected in all the samples that were imported within the indicated date range for the selected panel/panel group. The information that is displayed on this sub-tab is identical to the information that is displayed on the Variants sub-tab on the Patient tab. You can double-click a variant on this sub-tab to open the Variant tab. You can also right-click a variant on this sub-tab to open a context menu that has options that are identical to options that are on the context menu for the Variants sub-tab on the Patient tab. See “Patient tab, Variants sub-tab” on page 111.

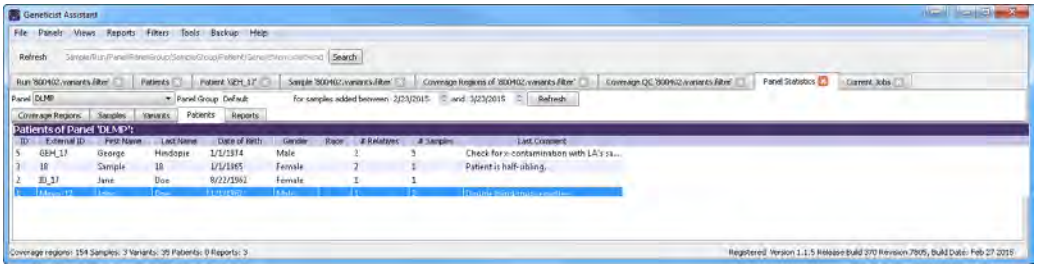
Figure 6-35: Panel Statistics tab, Variants sub-tab



Patients sub-tab (Panel Statistics tab via the Run tab)

The Patients sub-tab has a single pane, the Patients pane, that lists all the patients who were associated with all the samples that were imported within the indicated date range for the selected panel/panel group. You can double-click a patient in the pane to open the corresponding Patient tab. You can also right-click on a patient on this sub-tab to open a context menu with options that are identical to options that are on the context menu for the Patients tab. See "Accessing and Reviewing Patient Records" on page 95.

Figure 6-36: Panel Statistics tab, Patients sub-tab

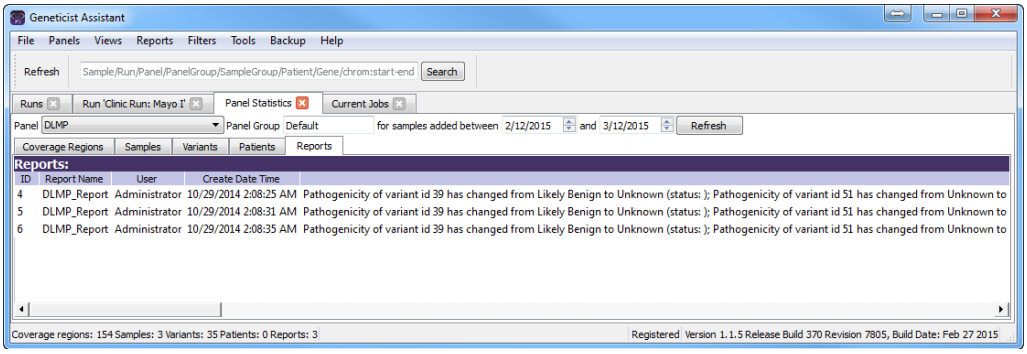


Reports sub-tab (Panel Statistics tab via the Run tab)

The Reports sub-tab contains a single pane, the Reports pane, that displays all the reports that were generated within the indicated date range for the selected panel/panel group and saved to the Geneticist Assistant database.

You can also select Reports > Saved Reports on the Geneticist Assistant main menu to open a Reports tab that displays these same reports.

Figure 6-37: Panel Statistics tab, Reports sub-tab



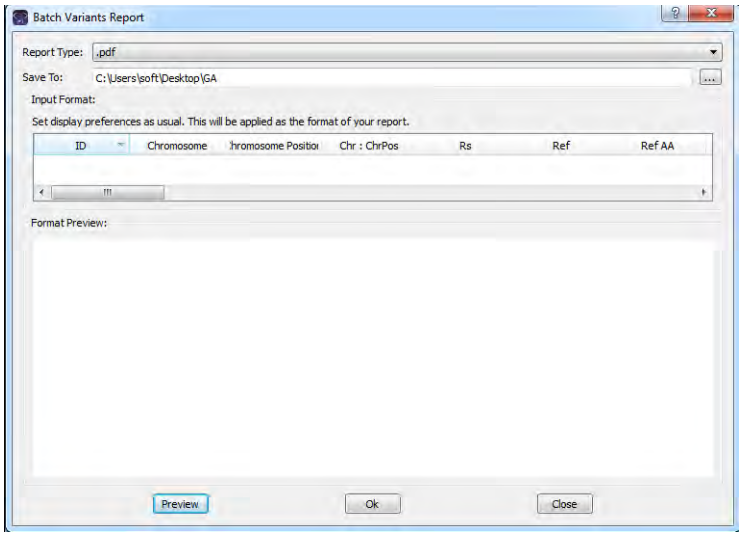
You can right-click anywhere on this sub-tab to open a context menu with options that are identical to the options that are on context menu for the Reports pane on the Patient sub-tab on the Patient tab. See "Patient tab, Patient sub-tab, Reports pane" on page 105.

For detailed information about generating and saving Panel Statistics reports, see Chapter 7, "Geneticist Assistant Reports," on page 203.

To manage a Batch Variants report (Run tab)

A *Batch Variants report* is a report that includes information for variants over a specified date range or other parameters. A *Batch Variants report configuration* determines the data that is to be included in the report, and if the report is to be available only to the user who created it, or to all users. The configuration does not include the report format (PDF, HTML and so on), or the location in which the report is to be saved. Instead, you must specify the format and location in which to save the report every time you generate a report for a new or saved configuration.

Figure 6-38: Batch Variants Report dialog box



In Geneticist Assistant, you can generate a Batch Variants report according to a new or saved configuration, modify a Batch Variants report configuration, or delete a Batch Variants report configuration. See:

- "To generate a Batch Variants report based on a new configuration" on page 167.
- "To generate a Batch Variants report based on a saved configuration" on page 168.
- "To modify a Batch Variants report configuration" on page 169.
- "To delete a Batch Variants report configuration" on page 170.

To generate a Batch Variants report based on a new configuration

When you generate a Batch Variants report, the report name is automatically assigned based on the variant ID for which you are generating the report (for example, MSH2_GC) and you cannot change this. You must select a location in which to save the report.

1. On the Batch Variants Report dialog box, on the Report Type dropdown list, select the report type. (The default value is PDF.)

2. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
   - Leave this location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Optionally, to view an onscreen preview of the report before it is generated and saved, click Preview.
4. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.
5. Click OK.

   The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

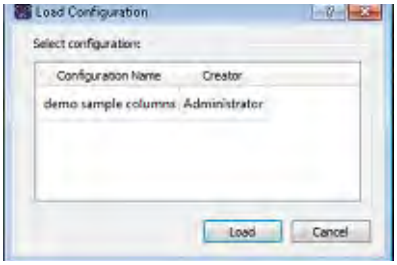
To generate a Batch Variants report based on a saved configuration

1. On the Batch Variants Report dialog box, select the output format for the report.
2. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
   - Leave the location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button ..., and then browse to and select the location in which the report is to be saved.
3. Right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.
4. Click Load Configuration.

   The Load Configuration dialog box opens. The dialog box displays all your private and public configurations as well as all the configurations that other users have made public.

Figure 6-39: Load Configuration dialog box



5. Select the configuration that you are loading, and then click Load.

   The Load Configuration dialog box closes. The Select Columns dialog box remains open. The pane display is updated dynamically after you load the configuration.

6. Select a configuration, and then click Load.

   The Load Configuration dialog box closes. The Select Columns dialog box remains open. The dialog box now reflects the columns that are to be included in the report based on the selected configuration.

7. Click Close (x) to close the Select Columns dialog box.

8. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.

9. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.

10. Click OK.

    The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To modify a Batch Variants report configuration

When you modify a Batch Variants report configuration, any reports that were generated according to the “old” (previous) configuration are not affected. Going forward, the report is generated based on the “new” configuration.

1. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank.

2. Do one of the following:

   - Leave this location as-is, and then go to Step 3.
   - To change this location, then next to the Save To field, click the Browse button  , and then browse to and select the location in which the report is to be saved.

3. Right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.

4. To modify the report configuration, see “To create/modify a pane configuration” on page 32.

5. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.

6. Click OK.

   The Batch Variants report is generated. After the report is successfully generated, a Batch Variants Report Finished dialog box opens, with a message that indicates where the report was saved.

7. Click OK.

   The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To delete a Batch Variants report configuration

You can delete a Batch Variants report configuration only if you created the report. When you delete a Batch Variants report configuration, any reports that were generated according to the "old" (previous) configuration are not affected. Going forward, the "old" configuration is simply not available for selection.

You are not prompted about confirming the deletion of a Batch Variants report configuration, so be very sure that the deletion must be deleted before doing so.

1. On the Batch Variants Report dialog box, right-click on any heading in the Input Format pane.

   The Select Columns dialog box opens.

2. See "To delete a pane configuration" on page 34.

3. Click Close (x) to close the Batch Variants Report dialog box.

To set or modify the Read/Write permissions for the samples in a run on a per sample basis

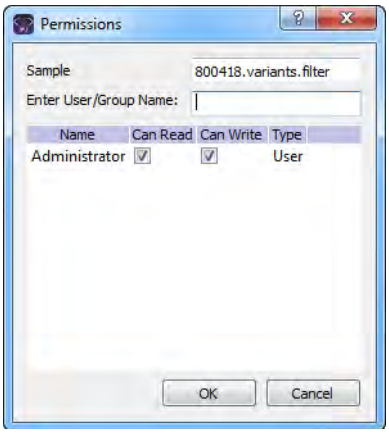
You can assign Read and Write permissions to all the samples in a run in a single step. See "To set or modify the Read/Write permissions for a run" on page 137.

1. If you have not already done so, open the Run tab.

2. Right-click the sample in the Samples pane, and on the context menu that opens, click Permissions.

   The Permissions dialog box opens. The first time that the dialog box opens, if the run was submitted using a Geneticist Assistant version with a build date of 4/8/15 or later, then the name of the user who submitted the run is displayed in the lower pane of the dialog box and both the Can Read and Can Write permissions are selected. If a standard user submitted the run, then the Administrator user name is also displayed. For runs that were submitted using earlier versions of Geneticist Assistant, the pane is blank. For subsequent openings, the dialog box displays the names of all the users and/or groups and their assigned permissions for the run. See Figure 6-40 on page 171.

Figure 6-40: Permissions dialog box (Run tab, Samples pane)



3. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to Step 3.

You can add or remove the Can Write permission for a user or group, or you can remove both the Can Read and Can Write permissions. Removing the Can Read permission but leaving the Can Write permission selected for a user or group has no effect.

4. Enter a user name (the login name for the user), or to assign the same permissions to multiple users in a single step, enter a group name.

   As you enter a user name or a group name, a dropdown list opens. The list is dynamically updated with user names or group names that are in your Geneticist Assistant database and that match the search string that you are entering.

5. Select the appropriate user name or group name from the list, and then press [ENTER].

   The user name or group name is displayed in the lower pane of the dialog box. The Read permission is already assigned for the selected user or group. If just this permission is selected, then the user or group can simply view the sample record. They cannot modify it.

6. Optionally, if the user or group must be able to modify the sample record as well, then select the Can Write permission.

7. Click OK.

Run's Status Changes pane

The Run's Status Changes pane shows the history of all the status changes that have been made for the run in chronological order. A New status entry is always displayed for a run. You can right-click anywhere on this pane to open a context menu with options:

Option	Description
Add All to Custom Report	Selects all the information that is displayed for all the statuses for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the sub-tab. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Reports pane (Run tab)

The Reports pane displays all the reports that were generated for the selected run and saved to the Geneticist Assistant database.

You can also select Reports > Saved Reports on the Geneticist Assistant main menu to open a Reports tab that displays these same reports.

You can right-click anywhere on this pane to open a context menu with options that are identical to the options that are on context menu for the Reports pane on the Patient sub-tab on the Patient tab. See "Patient tab, Patient sub-tab, Reports pane" on page 105.

For detailed information about generating and saving both standard and custom Run reports, see Chapter 7, "Geneticist Assistant Reports," on page 203.

The Sample Tab

To open the Sample tab, do one of the following:

- Right-click a sample entry on the Run tab, and on the context menu that opens, select Sample Details.
- Double-click a sample entry on the Run tab.

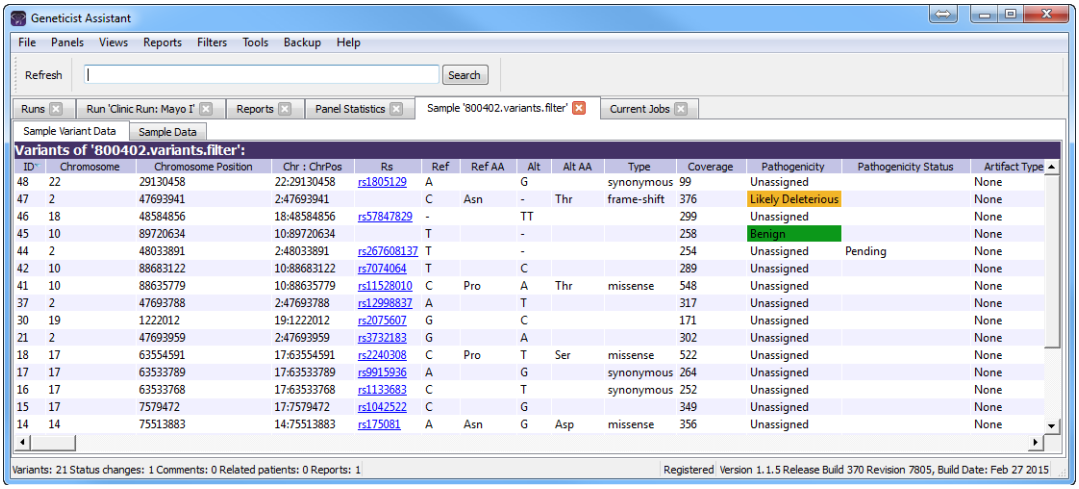
The Sample tab has two sub-tabs:

- The Sample Variant Data sub-tab. See "Sample Variant Data sub-tab" below.
- The Sample Data sub-tab. See "Sample Data sub-tab" on page 185.

Sample Variant Data sub-tab

The Sample Variant Data sub-tab is the tab the opens after you open the Sample tab. .

Figure 6-41: Sample tab, Sample Variant Data sub-tab



ID	Chromosome	Chromosome Position	Chr : ChrPos	Rs	Ref	Ref AA	Alt	Alt AA	Type	Coverage	Pathogenicity	Pathogenicity Status	Artifact Type
48	22	29130458	22:29130458	rs1805129	A		G		synonymous	99	Unassigned		None
47	2	47693941	2:47693941		C	Asn	-	Thr	frame-shift	376	Likely Deleterious		None
46	18	48584856	18:48584856	rs57847829	-		TT			299	Unassigned		None
45	10	89720634	10:89720634		T		-			258	Benign		None
44	2	48033891	2:48033891	rs267608137	T		-			254	Unassigned	Pending	None
42	10	88683122	10:88683122	rs7074064	T		C			289	Unassigned		None
41	10	88635779	10:88635779	rs11528010	C	Pro	A	Thr	missense	548	Unassigned		None
37	2	47693788	2:47693788	rs12998837	A		T			317	Unassigned		None
30	19	1222012	19:1222012	rs2075607	G		C			171	Unassigned		None
21	2	47693959	2:47693959	rs3732183	G		A			302	Unassigned		None
18	17	63554591	17:63554591	rs2240308	C	Pro	T	Ser	missense	522	Unassigned		None
17	17	63533789	17:63533789	rs9915936	A		G		synonymous	264	Unassigned		None
16	17	63533768	17:63533768	rs1133683	C		T		synonymous	252	Unassigned		None
15	17	7579472	17:7579472	rs1042522	C		G			349	Unassigned		None
14	14	75513883	14:75513883	rs175081	A	Asn	G	Asp	missense	356	Unassigned		None

The Sample Variant Data sub-tab has a single pane, the Variants pane, that displays information about *all* the variants that were identified in the selected sample. The pane lists the same information for each variant that was displayed on the Variants sub-tab on the Coverage QC tab. (See "Variants sub-tab (Coverage QC tab)" on page 148.) It also lists the following information if a sample group was used for the run and statistics were calculated for the sample group.

To calculate the statistics for the sample group, use the Manage Sample Group Statistics option on the column header context menu for the sub-tab. See "Manage Sample Group Statistics Columns" in Table 6-8 on page 174.

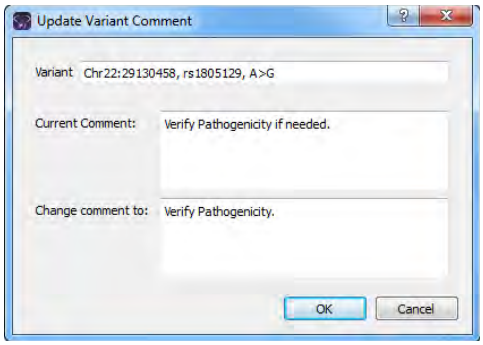
Table 6-8: Sample Group Statistics Columns

Column	Description
Number of Samples	Number of samples that.have been submitted for the sample group to which this sample is assigned.
Number of Samples Having the Variant	Number of samples in the group that contained the variant.
Coverage Min	The minimum coverage (number of reads) for the variant in the sample group.
Coverage Max	The maximum coverage (number of reads) for the variant in the sample group.
Coverage Median	The median coverage (number of reads) for the variant in the sample group.
Coverage Standard Deviation	The variation from the Coverage mean for the variant in the sample group.
Variant Frequency Min	The minimum frequency at which the variant was detected in the sample group. Any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.
Variant Frequency Max	The maximum frequency at which the variant was detected in the sample group. Any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.
Variant Frequency Median	The median frequency at which the variant was detected in the sample group. Any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.
Variant Frequency Mean	The mean frequency at which the variant was detected in the sample group. Any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.
Variant Frequency Standard Deviation	The variation from the Frequency mean for the variant in the sample group.
Read Balance Min	The minimum value for the balance in forward and reverse oriented reads with the variant in the sample group as reported in the VCF file.
Read Balance Max	The maximum value for the balance in forward and reverse oriented reads with the variant in the sample group as reported in the VCF file.
Read Balance Median	The median value for the balance in forward and reverse oriented reads with the variant in the sample group as reported in the VCF file.
Read Balance Mean	The mean value for the balance in forward and reverse oriented reads with the variant in the sample group as reported in the VCF file.
Read Balance Standard Deviation	The variation from the Read Balance mean for the variant in the sample group.

You can do the following for a variant that is displayed on the sub-tab:

- You can specify how the information about the variants is to be displayed on this sub-tab. See "To specify variant preferences" on page 53.
- You can select a different transcript for the variant on the Transcript dropdown list.
- You can single click twice in the Variant Comment field to select the current comment in the field and edit it, or if the field is blank, enter a new comment. After you add or edit a comment, and then click out of the field, the Update Variant Comment dialog box opens. See Figure 6-42 on page 175.

Figure 6-42: Update Variant Comment dialog box



The dialog box displays the variant for which you are adding or editing the comment, the current (old) comment, and the new comment. You can make additional edits to the new comment needed, and then click OK to close the dialog box and save the new or modified comment for the variant.

You can also use the Add Variant Comment option that is available on the context menu for the variant to add a new variant comment. See "Add Variant Comment" on page 177.

- You can click once in the Type field, the Zygosity field, the HGVS Genomic field, the HGVS Coding field, the HGVS Protein field, and/or the Read Balance field to select the current value in the field and edit it, or if the field is blank, enter a new value.

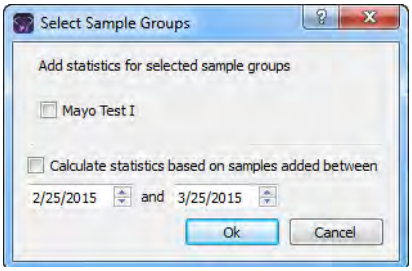
If you make a change to any of these values for the variant, then these changes are recorded for the variant on the Variant Changes pane on the Variant tab. See "Variant Changes pane" on page 196. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in the Geneticist Assistant application.

- You can right-click any column header to open a context menu with the following options:

Table 6-9: Sample tab, Sample Variant Data sub-tab column header context menu

Option	Description
Rename Header	Every data column is titled with a default column header. To rename a column header to better suit your working needs, right-click the column header, and on the context menu that opens, click Rename Header. The column header is selected, and you can then modify the column header as needed. See "Rename column headers" on page 35.

Table 6-9: Sample tab, Sample Variant Data sub-tab column header context menu (Continued)

Option	Description
Manage Sample Group Statistics Columns	Opens the Select Sample Groups dialog box. • If you used sample groups for a run, then you can indicate the date range for which to calculate statistics based on the added samples. • If you did not use sample groups for a run, you can clear the options for selected sample groups, which hides the corresponding Sample Group Statistics columns from the sub-tab display. *Figure 6-43: Select Sample Groups dialog box* 
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the sub-tab context menu. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations. **Note:** If you set column display preferences here, then you can apply these same display preferences for variants in all the variant sub-tabs and panes (with the exception of the Sample Group columns on the Sample Variant Data sub-tab on the Sample tab and the Variant pane on the Variant tab) in a single step. See "To apply shared preferences or to clear all tab preferences" on page 51.

- You can right-click a variant in the pane to open a context menu with the following options:

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu

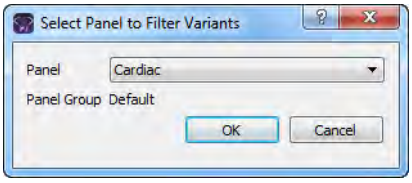
Option	Description
Variant Details	Opens the Variant tab, which displays detailed information about the variant. See "The Variant Tab" on page 190.
Show Variants Filtered by Panel	Displays all the variants in the selected sample filtered by a selected panel. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected run. *Figure 6-44: Select Panel to Filter Variants dialog box* 

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu (Continued)

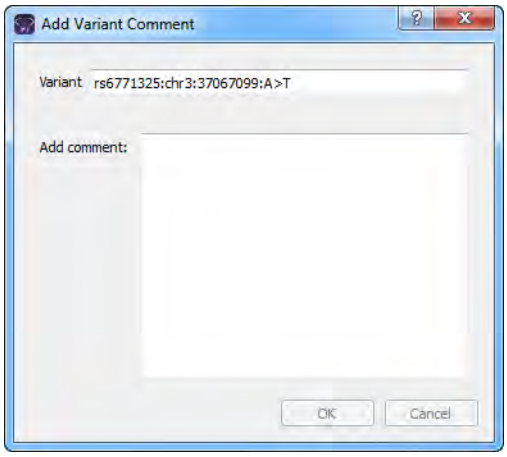
Option	Description
Edit Variant - Contains all the sub-menu options for editing and deleting a variant.	
Add Variant Comment	Opens the Add Variant Comment dialog box. You can add a comment for the variant in the Add Comment field, and then click OK. The comment is displayed in the Variant Comment field in the Variants pane on the Search Results tab, the Variant Comment field in the Variants sub-tab on the Coverage QC tab, the Variant Comment field on the Sample Variant Data sub-tab on the Sample tab, the Variant Comment field in the Variant pane on the Variant tab, and the Variant Comments pane on the Variant tab. *Figure 6-45: Add Variant Comment dialog box*  **Note:** If multiple comments have been added for a variant, then with the exception of the Variant Comments pane on the Variant tab, only the last added comment is displayed in all the locations that are detailed above. All the comments that have been added for a variant, including the last added, are displayed in reverse chronological order in the Variant Comments pane on the Variant tab.

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu (Continued)

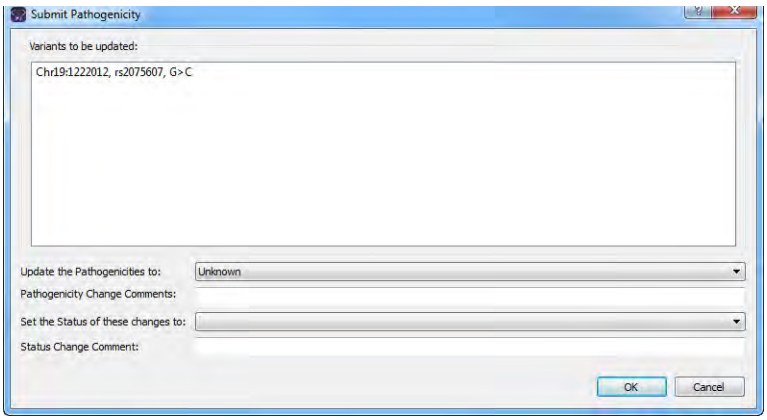
Option	Description
Update Pathogenicity	Opens the Submit Pathogenicity dialog box. You can update the pathogenicity for the selected variant and/or the status of the pathogenicity in this dialog box. You can also add a comment about the pathogenicity change and/or pathogenicity status change. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be; for example, Deleterious. (If you cannot determine the pathogenicity of the variant even after reviewing all the relevant information, you can set the status to Unknown.) After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant. *Figure 6-46: Submit Pathogenicity dialog box*  **Note:** You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box.You can also press and hold the CTRL key to select multiple variants for updating the pathogenicities and the pathogenicity statuses at the same time. **Note:** You can also update the Pathogenicity and the Pathogenicity Status for a variant on the Variants sub-tab on the Patient tab and the Variants sub-tab on the Coverage QC tab. The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See “Managing Objects” on page 41. • Optionally, you can add supporting information for this pathogenicity change. See “To add supporting information for a pathogenicity change” on page 198. • You always have the option of updating just the pathogenicity status for a variant at a later date. See “Update Pathogenicity Status” below.

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu (Continued)

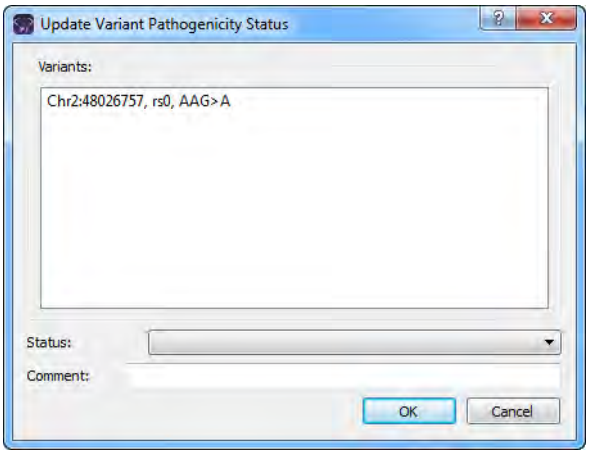
Option	Description
Update Pathogenicity Status	Opens the Update Variant Pathogenicity Status dialog box. You can update the pathogenicity status for the variant in this dialog box. You can also add a comment for the variant. *Figure 6-47: Update Variant Pathogenicity Status dialog box*  **Note:** By default, only two statuses, Pending or Confirmed, are available. You have the option of adding custom pathogenicity statuses. See "Managing Objects" on page 41. **Note:** You can also update the Pathogenicity Status for a variant on the Variants sub-tab on the Patient tab and the Variants sub-tab on the Coverage QC tab. **Note:** You can also double-click in the Pathogenicity Status column for a variant to open the Update Variant Pathogenicity Status dialog box. You can also press and hold the CTRL key to select multiple variants for updating the pathogenicity statuses at the same time.

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu (Continued)

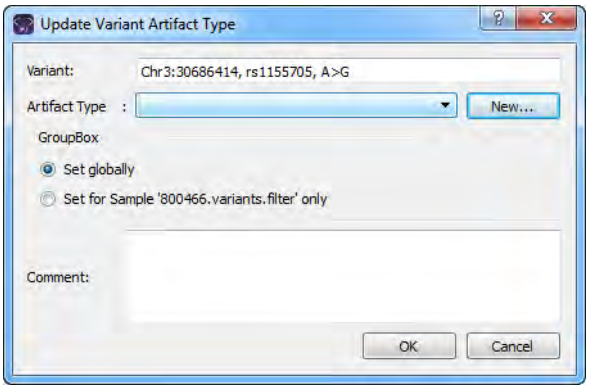
Option	Description
Update Artifact Type	Opens the Update Artifact Type dialog box. You can specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type. *Figure 6-48: Update Variant Artifact Type dialog box*  When you specify the artifact type, you can set the artifact type as the following: • Set globally—Selected by default. Set this artifact type for the variant, regardless of the sample in which the variant is detected. • Set for Sample—Set this artifact type for this variant only for the indicated sample. You also have the option of adding comments for the variant artifact type. The following information is relevant to updating the artifact type for a variant: • You can press and hold the CTRL key to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant on the Variants sub-tab on the Patient tab, on the Variants sub-tab on the Coverage QC tab, or on the Variants sub-tab on the Panel Statistics tab via the Run tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See “Managing Objects” on page 41.
Delete Variant	Deletes the selected variant from the sample.
External - Contains all the sub-menu options for viewing the variant in third-party tools.	
View in Alamut	Opens the variant for viewing in Alamut. **Note:** To view a variant in Alamut, your Alamut settings must be specified. See “To specify Alamut settings” on page 54.

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu (Continued)

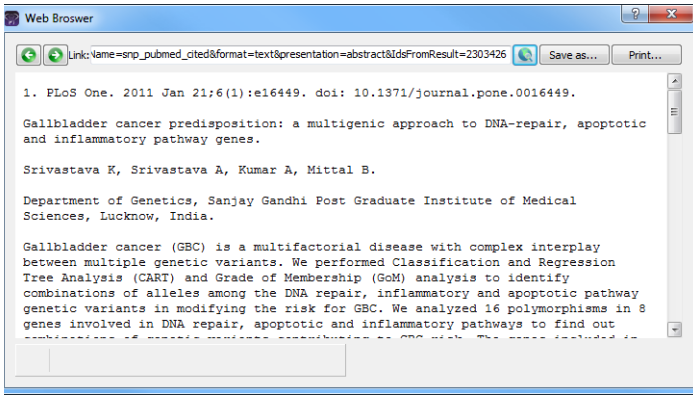
Option	Description
View PubMed Abstract	Searches PubMed for the variant and displays the appropriate abstract in the PubMed window. *Figure 6-49: PubMed window*  You can do the following for the abstract: • In the Link field, enter a different PubMed URL, and then click the Go To icon to navigate to abstract. • Click Save As to save the abstract as a PDF with a name and in a location of your choosing. • Click Print to print the abstract.
Export - Contains all the sub-menu options for exporting a variant.	
Export Selected Variants to VCF	Exports the selected variants to a VCF file. Press and hold the CTRL key to select multiple variants. You must specify both the file name and the location in which to save the file.
Export All Variants to VCF	Exports all the variants to a VCF file. You must specify both the file name and the location in which to save the file.
Reports - Contains all the sub-menu options for generating a report that includes the variant.	
Add Selected Variants to Action List	Adds the selected variant to an Action List. Press and hold the CTRL key to select multiple variants. See "To add selected panel regions or variants to an Action list" on page 182.
Add Selected Variants to Custom Report	Selects all the data that is displayed for the selected variants for inclusion in a custom report. Press and hold the CTRL key to select multiple variants. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Add All Variants to Custom Report	Selects all the data that is displayed for all the variants for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.

Table 6-10: Sample tab, Sample Variant Data sub-tab context menu (Continued)

Option	Description
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the sub-tab. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations. **Note:** If you set column display preferences here, then you can apply these same display preferences for variants in all the variant sub-tabs and panes (with the exception of the Sample Group columns on the Sample Variant Data sub-tab on the Sample tab and the Variant pane on the Variant tab) in a single step. See "To apply shared preferences or to clear all tab preferences" on page 51.

To add selected panel regions or variants to an Action list

An *Action list* is a list of items that require some type of follow up. You can create multiple action lists relative to the type of follow up that is needed. You can create an Action List for panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab or for variants on the Variants sub-tab on the Coverage QC tab or the Sample Variant Data sub-tab on the Sample tab. For example, if several panel regions failed the coverage settings for a run sample, then you can make an Action List that consists of these regions and indicate that you want to review the coverage values for these regions.

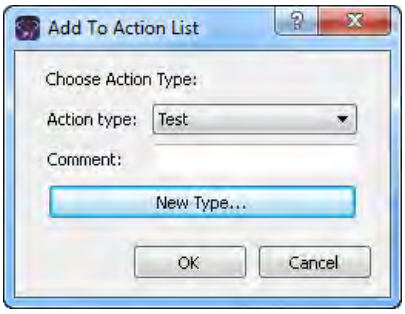
1. Do one of the following:

   - To select a single panel region for the Action list, right-click the appropriate panel region on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab, and on the context menu that opens, click Add Selected Regions to Action List.
   - To select multiple panel regions, press and hold the CTRL key as you select the appropriate panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab, and then right-click any selected panel region, and on the context menu that opens, click Add Selected Regions to Action List.
   - To select a single variant for the Action list, right-click on the appropriate variant on the Variants sub-tab on the Coverage QC tab or the Sample Variant Data sub-tab on the Sample tab, and on the context menu that opens, click Add Selected Variants to Action List.
   - To select multiple variants, press and hold the CTRL key as you select the appropriate variants on the Variants sub-tab on the Coverage QC tab or the Sample Variant Data sub-tab on the Sample tab, and then right-click on any selected variant, and on the context menu that opens, click Add Selected Variants to Action List.

   The Add To Action List dialog box opens. If the Action Type that you want to use is already available on the Action type dropdown list, then go to Step 5; otherwise, if this is the first time that you are creating an Action List for the pane or the Action Type that you

want to use is not available on the Action Type dropdown list, then you must add at least one type to the list. Go to Step 2.

Figure 6-50: Add To Action List dialog box



2. Click New Type.

   The Add To Action List dialog box is refreshed with an option for adding an action type.

3. In the New Type field, enter an action type (for example, Review Coverage), and then click Add.

   The action type is added to the Action Type list. The Add to Action List dialog box is refreshed to show the full Action Type dropdown list and Comment field.

4. Repeat Step 2 and Step 3 as needed to add more action types.

5. On the Action Type dropdown list, select the appropriate action type.

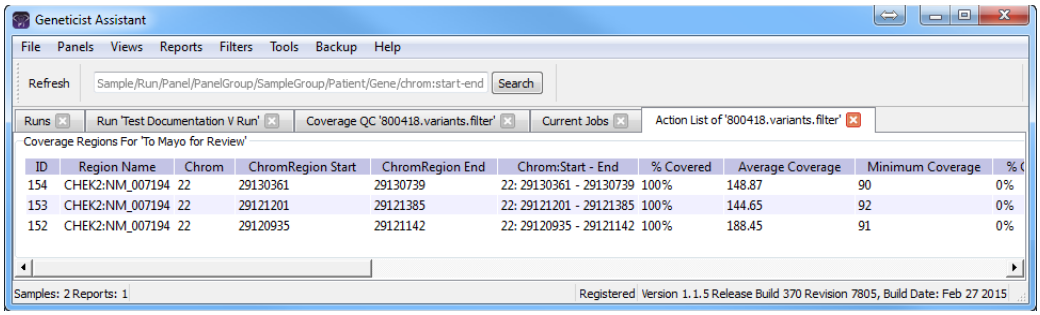
If you are creating an Action List for panel regions, then only the action types that were created specifically for panel regions are available. Likewise, if are creating an Action List for variants, then only the action types that were created specifically for variants are available.

6. Optionally, in the Comment field, enter a comment that explains/supports your decision for adding the selected panel regions or variants to an Action list.

7. Click OK.

   The Add To Action List dialog box closes. The Action List tab for the sample opens. Different panes (grouped by Action Type) list any panel regions for the sample that have been selected for an Action list and any variants for the sample that have been selected for an Action list. See Figure 6-51 on page 184.

Figure 6-51: Action List tab

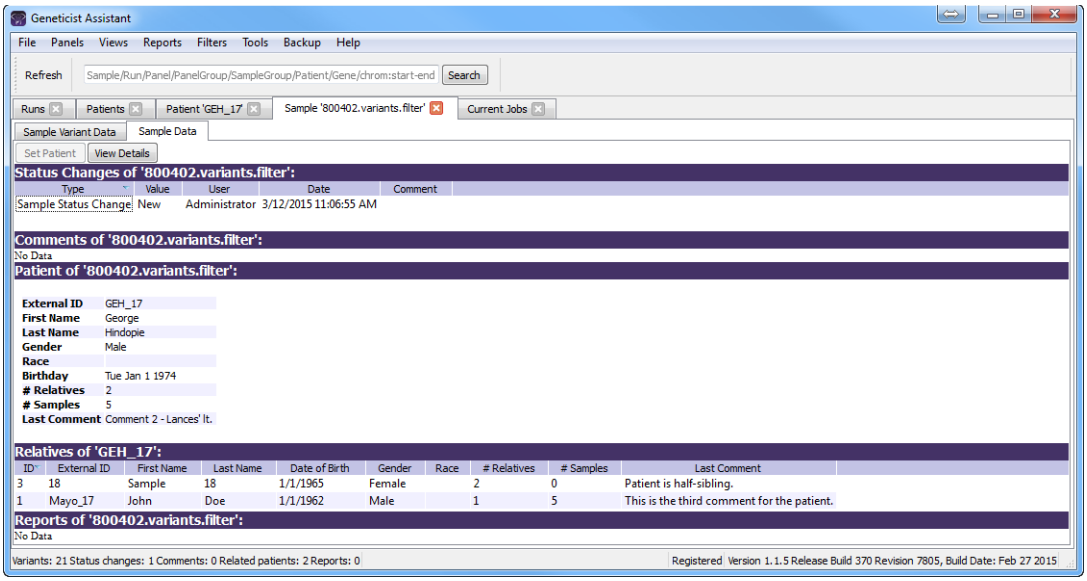


To save an Action List, you can generate a standard Action List Report. If multiple panes are included on the Action List tab, then by default, all these panes are included in the report. To remove a pane from the report, before generating the report, right-click on the pane that you want to remove, and then click Remove.
See "Generating a Standard Report" on page 205.

Sample Data sub-tab

The Sample Data sub-tab contains five panes—Status Changes, Comments, Patient, Related Patients, and Reports—that provide different information about the sample.

Figure 6-52: Sample tab, Sample Data sub-tab



A context menu is available for each pane. See:

- "Status Changes pane" below.
- "Comments pane (Sample Data sub-tab on the Sample tab)" on page 186.
- "Patient pane (Sample Data sub-tab for the Sample tab)" on page 187.
- "Related Patients pane" on page 188.
- "Reports pane (Sample data sub-tab on the Sample tab)" on page 189.

Status Changes pane

The Status Changes pane lists an entry for each time the status was changed for the sample. Each entry details the value for the sample status, the name of the user who changed the sample status, the date and time of the status change, and any comments that the user entered in support of the change. The pane always contains an entry for the initial submission of the sample with the status of the sample set to New.

Remember, you can change the status for a sample on the Run tab. See "To set the sample status" on page 140.

Comments pane (Sample Data sub-tab on the Sample tab)

The Comments pane displays all the comments that have been added for the sample in reverse chronological order. You can-right click a comment in the pane to open a context menu with the following options:

Table 6-11: Sample tab, Sample Data sub-tab, Comments pane context menu

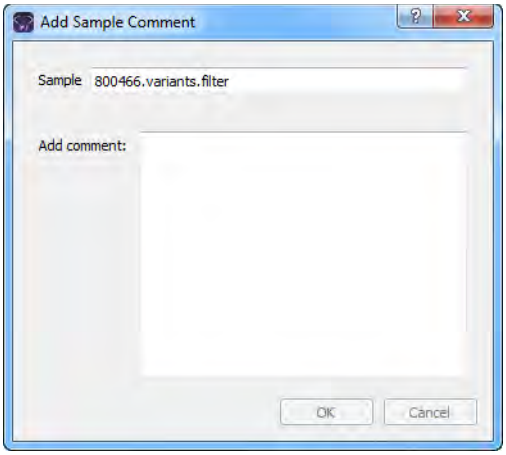
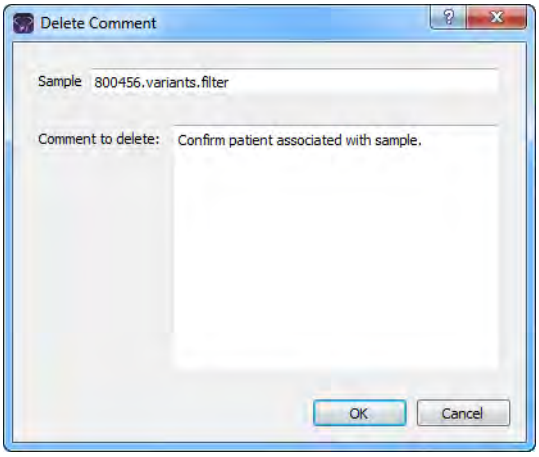
Option	Description
Add Sample Comment	Opens the Add Sample Comment dialog box. You add a comment for the sample in the Add Comment field. *Figure 6-53: Add Sample Comment dialog box*  **Note:** If multiple comments have been added for a sample, then these comments are displayed in reverse chronological order in the Comments pane on this tab. Only the last added comment is displayed in the Comments field in the Samples pane on the Samples data sub-tab on the Patient tab and in the Samples pane on the Run tab.
Delete Sample Comment	Opens the Add Delete Comment dialog box. Click OK to delete the comment for the sample and close the dialog box. *Figure 6-54: Delete Comment dialog box* 

Table 6-11: Sample tab, Sample Data sub-tab, Comments pane context menu (Continued)

Option	Description
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the sub-tab. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Patient pane (Sample Data sub-tab for the Sample tab)

The Patient pane displays information about the patient who is associated with the sample. If a patient is not associated with the sample, then Set Patient (displayed above the Status Changes pane) is available and you can associate one. If a patient is associated with the sample, then View Details (displayed above the Status Changes pane) is available and you can view details about the associated patient. You can also right-click anywhere on the information for the associated patient to open a context menu with the following options:

Table 6-12: Sample tab, Sample Data sub-tab, Patient pane context menu

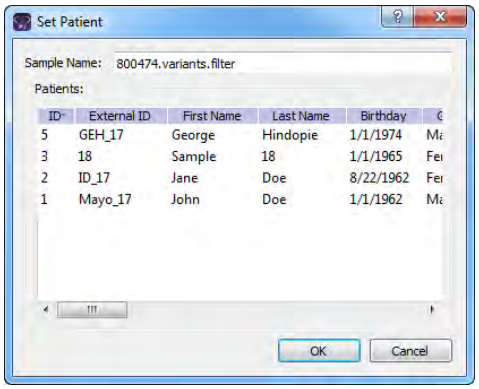
Option	Description
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Add Patient External ID to Custom Report	Selects only the External ID for the patient for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.

To associate a patient with a sample

1. If you have not already done so, do one of the following to open the Sample tab for the sample:
   - Right-click a sample entry on the Run tab, and on the context menu that opens, select Sample Details.
   - Double-click a sample entry on the Run tab.
2. Open the Sample Data sub-tab.
3. At the top of the sub-tab, click Set Patient.

   The Set Patient dialog box opens. The dialog box lists all the patients in your Geneticist Assistant database. See Figure 6-55 on page 188.

Figure 6-55: Set Patient dialog box



4. Select the patient, and then click OK.

   The Set Patient dialog box closes. The patient information is displayed in the Patient pane.

To view details about an associated patient

1. If you have not already done so, do one of the following to open the Sample tab for the sample:
   - Right-click a sample entry on the Run tab, and on the context menu that opens, select Sample Details.
   - Double-click a sample entry on the Run tab.
2. Open the Sample Data sub-tab.
3. At the top of the sub-tab, click View Details.

   The Patient tab opens.
4. Continue to "Accessing and Reviewing Patient Records" on page 95.

Related Patients pane

The Related Patients pane displays all the relatives of the patient who is associated with the sample. Right-click a patient in the Related Patients pane to open a context menu with the following options:

Table 6-13: Sample tab, Related Patient pane context menu

Option	Description
Patient Details	Opens the Patient tab for the selected related patient. All the options on the Patient tab are now available for the related patient. See "Accessing and Reviewing Patient Records" on page 95. **Note:** You can also double-click a patient entry to open the Patient tab.

Table 6-13: Sample tab, Related Patient pane context menu (Continued)

Option	Description
Add New Relationships	The Add New Patient dialog box opens. By default, the Is Related to Existing Patient option is selected and unavailable. Add the needed relationships for the patient. See Step 3 of "To add new patients one at a time" on page 118.
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Reports pane (Sample data sub-tab on the Sample tab)

The Reports pane displays all the reports that were generated for the selected sample and saved to the Geneticist Assistant database.

You can also select Reports > Saved Reports on the Geneticist Assistant main menu to open a Sample Reports tab that displays these same reports.

You can right-click anywhere on this sub-tab to open a context menu with options that are identical to the options that are on context menu for the Reports pane on the Patient sub-tab on the Patient tab. See "Patient tab, Patient sub-tab, Reports pane" on page 105.

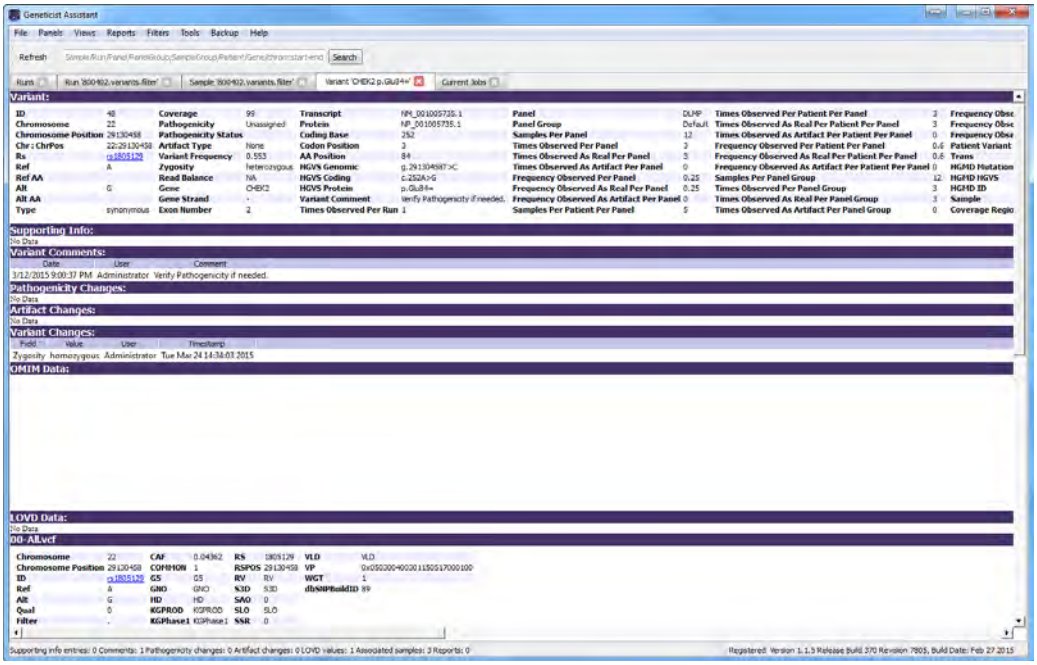
For detailed information about generating and saving both standard and custom Sample reports, see Chapter 7, "Geneticist Assistant Reports," on page 203.

The Variant Tab

The Variant tab displays detailed information for the variant that was selected on the Sample tab. To open the Variant tab, do one of the following:

- Double-click a variant on the Variants sub-tab on the Patient tab.
- Double-click a variant on the Variant sub-tab on the Panel Statistics tab.
- Right-click a variant on the Sample Variant Data sub-tab on the Sample tab, and on the context menu that opens, select Variant Details.
- Double-click a variant on the Sample Variant Data sub-tab on the Sample tab.

Figure 6-56: Variant tab



The Variant tab can contain up to 15 panes. Each pane provides different information about the variant. A context menu is available for each pane, and some of the menus have options that are unique to the pane. See:

- “Variant pane” on page 192.
- “Supporting Information pane” on page 193.
- “Variant Comments pane” on page 194.
- “Historical Pathogenicity Changes pane” on page 195.
- “Artifact Changes pane” on page 195.
- “Variant Changes pane” on page 196.

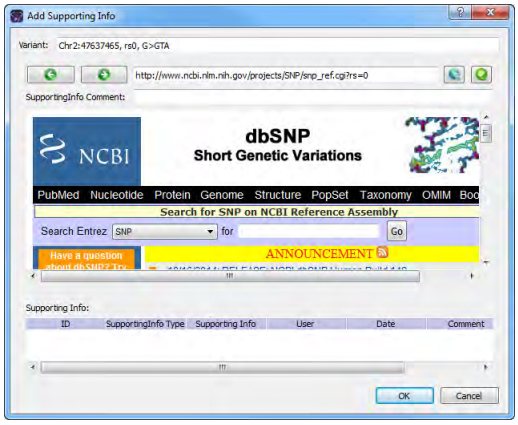
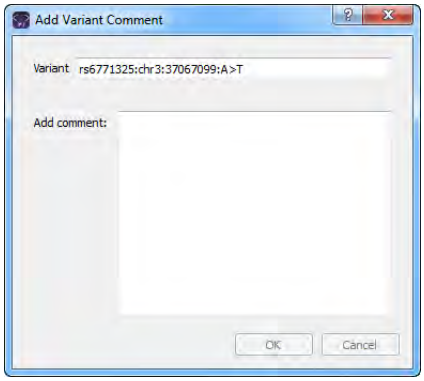
- "LOVD Data pane" on page 196.
- "dbSNP (*/00-All.vcf) pane" on page 196.
- "COSMIC (*/CosmicCodingMuts_*_noLimit.vcf) pane" on page 197.
- "Exome Variant Server (*/ESP*.vcf) pane" on page 197.
- "NIST (*/NISTIntegratedCalls_*.vcf) pane" on page 197.
- "ClinVar (*/clinvar_00-latest.vcf) pane" on page 198.
- "dbNSFP pane (Variant Prediction)" on page 198.
- "Associated Samples pane" on page 200.
- "Variant's Reports pane" on page 201.

If any of the Reference panes are not displayed, contact SoftGenetics to download the latest reference files.

Because so many panes can be displayed on the Variant tab, remember, you can always select which panes to show and which to hide to make it easier for you to review variant data. See "Show/hide a tab pane" on page 27.

Variant pane

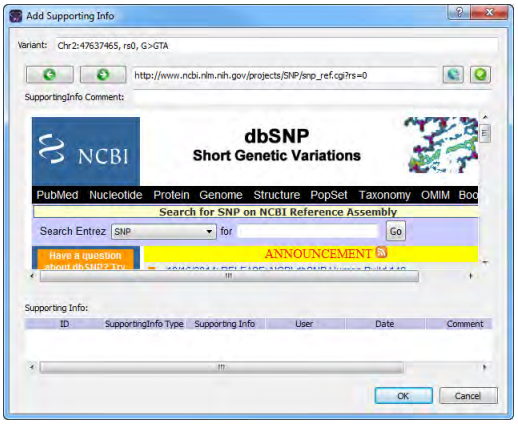
The Variant pane is a read-only pane that lists all the same information that is displayed by default for the variant on the Variants sub-tab on the Coverage QC tab. (See "Variants sub-tab (Coverage QC tab)" on page 148.) You can right-click the variant to open a context menu with the following options:

Option	Description
Submit Supporting Hyperlink	Opens the Add Supporting Info dialog box, which, by default, displays the dbSNP web page for the variant. You can use the browser in this dialog box to select this web page as supporting information for the variant, or navigate to a different page. *Figure 6-57: Add Supporting Info dialog box*  **Note:** See Step 9 of "To add supporting information for a pathogenicity change" on page 198.
Add Comment	Opens the Add Variant Comment dialog box. You can add a comment for the variant in the Add Comment field, and then click OK. The comment is displayed in the Variant Comment field in the Variant pane on the Variant tab, the Variant Comments pane on the Variant tab, the Variant Comment field in the Variants pane on the Search Results tab, the Variant Comment field on the Variants sub-tab on the Patient tab, the Variant Comment field on the Variants sub-tab on the Coverage QC tab, and the Variant Comment field on the Sample Variant Data sub-tab on the Sample tab. *Figure 6-58: Add Variant Comment dialog box* 

Option	Description
Note: If multiple comments have been added for a variant, then with the exception of the Variant Comments pane on the Variant tab, only the last added comment is displayed in all the locations that are detailed above. All the comments that have been added for a variant, including the last added, are displayed in reverse chronological order in the Variant Comments pane on the Variant tab.	

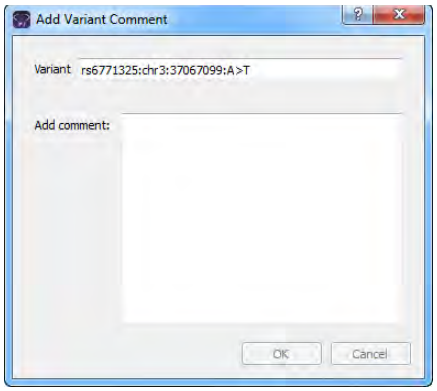
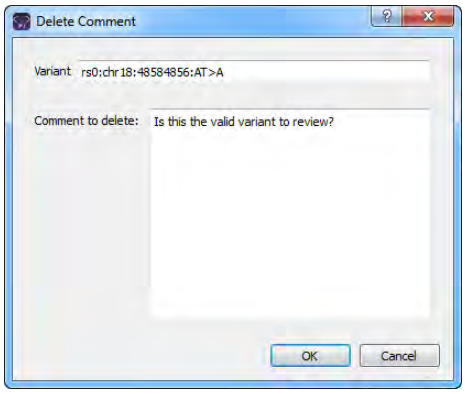
Supporting Information pane

The Supporting Information pane displays all the reference information and/or links that you have submitted as supporting information for a variant and/or a pathogenicity change for the variant. (See "To add supporting information for a pathogenicity change" on page 198.) You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit Supporting Hyperlink	Opens the Add Supporting Info dialog box, which, by default, displays the dbSNP web page for the variant. You can use the browser in this dialog box to select this web page as supporting information for the variant, or navigate to a different page. *Figure 6-59: Add Supporting Info dialog box*  **Note:** See Step 9 of "To add supporting information for a pathogenicity change" on page 198.
Add All to Custom Report	Selects all the information that is displayed for all supporting information for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Variant Comments pane

The Variant Comments pane displays all the comments that have been added for the variant in reverse chronological order. You can right-click a comment in the pane to open a context menu with the following options:

Option	Description
Add Comment	Opens the Add Variant Comment dialog box. You can add a comment for the variant in the Add comment field, and then click OK. The comment is displayed in the Variant Comment field in the Variant pane on the Variant tab, the Variant Comments pane on the Variant tab, the Variant Comment field in the Variants pane on the Search Results tab, the Variant Comment field on the Variants sub-tab on the Patient tab, the Variant Comment field on the Variants sub-tab on the Coverage QC tab, and the Variant Comment field on the Sample Variant Data sub-tab on the Sample tab. *Figure 6-60: Add Variant Comment dialog box*  **Note:** If multiple comments have been added for a variant, then with the exception of the Variant Comments pane on the Variant tab, only the last added comment is displayed in all the locations that are detailed above. All the comments that have been added for a variant, including the last added, are displayed in reverse chronological order in the Variant Comments pane on the Variant tab.
Delete Comment	Opens the Delete Comment dialog box. Click OK to delete the comment for the variant and close the dialog box. *Figure 6-61: Delete Comment dialog box* 

Option	Description
Add All To Custom Report	Selects all the information that is displayed for all comments for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See “Work with columns in a tab pane” on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Historical Pathogenicity Changes pane

The Historical Pathogenicity Changes pane displays the history of the changes that have been made to the pathogenicity for the variant in reverse chronological order. When the Variant tab first opens for a variant, the Pathogenicity is set to Unassigned and the Pathogenicity Status does not have a value. If the Pathogenicity is changed to Deleterious, and the Status is set to Confirmed, for example, then this change and the name of the user who made the change, and the date and the time of the change are recorded and displayed in this pane. You can right-click in the pane to open a context menu with the following options:

Option	Description
Add All To Custom Report	Selects all the information that is displayed for all pathogenicity changes for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See “Work with columns in a tab pane” on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Artifact Changes pane

The Artifact Changes pane displays the history of the changes that have been made to the artifact type for the variant in reverse chronological order. When the Variant tab first opens for a variant, the Artifact Type is set to None. If the Artifact Type is set to Sequencing, for example, then this change and the name of the user who made the change, and the date and the time of the change are recorded and displayed in this pane. You can right-click in the pane to open a context menu with the following options:

Option	Description
Add All To Custom Report	Selects all the information that is displayed for all artifact changes for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.

Option	Description
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. This option is also available on the column header context menu for the pane. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

Variant Changes pane

The Artifact Changes pane displays the history of the changes that have been made to the variant in reverse chronological order, for example, a change in the zygosity of the variant from heterozygous to homozygous. The value that was changed for the variant, the name of the user who made the change, and the date and the time of the change are recorded and displayed in this pane. You can right-click in the pane to open a context menu with the following options:

Option	Description
Add All To Custom Report	Selects all the information that is displayed for all variant changes for inclusion in a custom report. See Chapter 7, "Geneticist Assistant Reports," on page 203.
Load Columns	Opens a sub-menu on which you can click Manually to open the Select Columns dialog box and select which columns to show and which to hide dynamically, or select a configuration from the list of available column configurations. **Note:** See "Work with columns in a tab pane" on page 30 for information about showing and hiding columns dynamically, and creating pane configurations.

LOVD Data pane

The LOVD Data pane displays the information from the LOVD (Leiden Open Variation Database) for the variant. You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" on page 198.
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

dbSNP (*/00-All.vcf) pane

The dbSNP pane displays the information from the dbSNP database for the variant. You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" on page 198.

Option	Description
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

COSMIC (*/CosmicCodingMuts_*_noLimit.vcf) pane

The COSMIC pane displays the information from the COSMIC database for the variant. You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" on page 198.
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

Exome Variant Server (*/ESP*.vcf) pane

The Exome Variant Server pane displays the information from the Exome Variant Server database for the NHLBI GO Exome Sequencing Project (ESP) for the variant. Right-click any field in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" on page 198.
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

NIST (*/NISTIntegratedCalls_*.vcf) pane

The NIST pane displays the information for the variant from the NIST whole human genome Reference Materials database for highly confident variant calls. You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" on page 198.
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

ClinVar (*/clinvar_00-latest.vcf) pane

The ClinVar pane displays the information for the variant from the ClinVar database. You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" on page 198.
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

dbNSFP pane (Variant Prediction)

The dbNSFP pane displays the information from the dbNSFP database. You can right-click in the pane to open a context menu with the following options:

Option	Description
Submit as Supporting Info	Submits the information in the field as supporting information for a variant. See "To add supporting information for a pathogenicity change" below.
Add value to variant table	Adds the selected field to the Variant pane, the Variants sub-tab on the Coverage QC tab, and the Variants pane on the Sample Variant Data sub-tab on the Sample tab.

To add supporting information for a pathogenicity change

You can add supporting information for a variant. Typically, this is information that you used to determine the appropriate pathogenicity for the variant. You can reference information from an external database or from a web page as supporting information. The supporting information is displayed in the Supporting Information pane on the Variant tab. (See "Supporting Information pane" on page 193.)

1. If you have not done so already, do one of the following to open the Variant tab:

- Double-click a variant on the Variants sub-tab on the Patient tab.
- Double-click a variant on the Variant sub-tab on the Panel Statistics tab.
- Right-click a variant on the Sample Variant Data sub-tab on the Sample tab, and on the context menu that opens, select Variant Details.
- Double-click a variant on the Sample Variant Data sub-tab on the Sample tab.

2. Do one of the following:
    - To reference information from any reference track, go to Step 3.
    - To reference a specific web page, go to Step 8.
3. In the appropriate reference information pane, review the information that is to be used for supporting information. See:
    - "LOVD Data pane" on page 196.

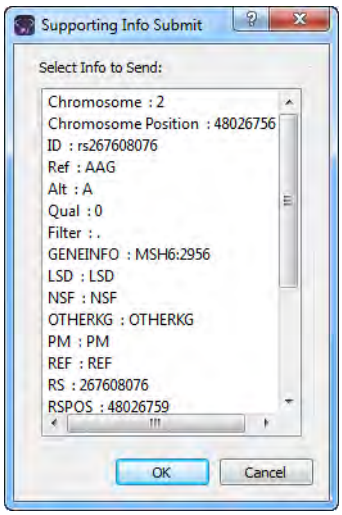
- "dbSNP (*/00-All.vcf) pane" on page 196.
- "COSMIC (*/CosmicCodingMuts_*_noLimit.vcf) pane" on page 197.
- "Exome Variant Server (*/ESP*.vcf) pane" on page 197.
- "NIST (*/NISTIntegratedCalls_*.vcf) pane" on page 197.
- "ClinVar (*/clinvar_00-latest.vcf) pane" on page 198.
- "dbNSFP pane (Variant Prediction)" on page 198.

This information is included in the References folder. Use the scroll bar at the bottom of the pane to view the available reference information for the variant. Note that all reference information is not available for all variants.

4. Right-click anywhere in a pane, and on the context menu that opens, click Submit As Supporting Info.

   The Supporting Info Submit dialog box opens. The dialog box lists all the reference information and the corresponding values that can be selected from the pane as supporting information.

Figure 6-62: Supporting Info Submit dialog box



5. Select the information that is to be used as supporting information, and then click OK.

   The Add Supporting Info dialog box closes. The information is displayed in the Supporting Information pane. See "Supporting Information pane" on page 193.

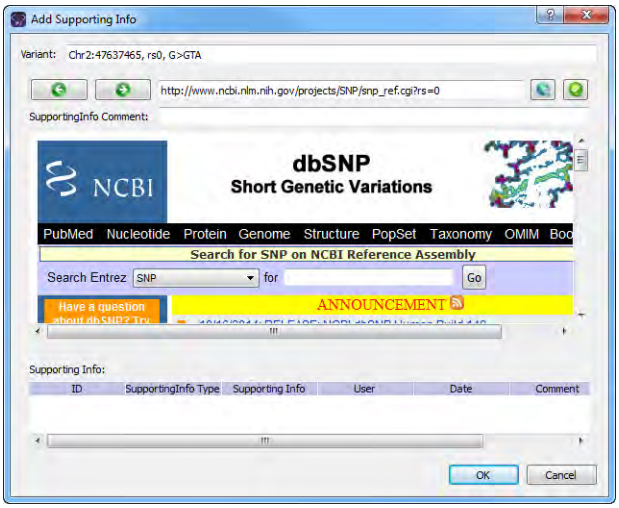
6. Repeat Step 3 through Step 5 for each piece of reference information that you want to add as supporting information.

7. Optionally, continue to Step 8 if you want to also add supporting information for the variant from a web page.

8. Right-click anywhere on the Variant pane or the Supporting Information pane, and on the context menu that opens, click Submit Supporting Hyperlink.

   The Add Supporting Info (for a web page) dialog box opens. By default, the dialog box displays the dbSNP page for the variant.

Figure 6-63: Add Supporting Info dialog box



9. Do one of the following:

   - Click the Use This Link icon to accept the NCBI dbSNP page. The URL is added under Supporting Info on the dialog box.
   - Enter a new URL, click the Go To icon to navigate to the web page, and then click Click the Use This Link icon to accept the web page. The URL is added under Supporting Info on the dialog box.

You can also use the Add Supporting Info dialog box like a standard web browser and search for the necessary URL.

10. Click OK.

    The Add Supporting Info dialog box closes. The URL is displayed in the Supporting Information pane on the Variant tab. See "Supporting Information pane" on page 193.

Associated Samples pane

The Associated Samples pane lists all the samples in which the variant has been identified. You can right-click in the pane to open a context menu with the following options:

Option	Description
Sample Detail	Opens the Sample tab for the sample. See "The Sample Tab" on page 173.

Option	Description
Add All to Custom Report	Selects all the information that is displayed for all associated samples for inclusion in a custom report. See Chapter 7, “Geneticist Assistant Reports,” on page 203.

Variant’s Reports pane

The Reports pane displays all the reports that were generated for the selected variant and saved to the Geneticist Assistant database.

You can also select Reports > Saved Reports on the Geneticist Assistant main menu to open a Reports tab that displays these same reports.

You can right-click anywhere on this sub-tab to open a context menu with options that are identical to the options that are on context menu for the Reports pane on the Patient sub-tab on the Patient tab. See “Patient tab, Patient sub-tab, Reports pane” on page 105.

For detailed information about generating and saving both standard and custom Run reports, see Chapter 7, “Geneticist Assistant Reports,” on page 203.

Chapter 6
Geneticist Assistant Data Review

Chapter 7
Geneticist Assistant Reports

As discussed in Chapter 6, “Geneticist Assistant Data Review,” on page 129, after a run is successfully completed, many options are available for reviewing the data on the various tabs and panes on the Geneticist Assistant main window. If you want to generate an output file of the data, then you have two options. You can generate a standard report, or you can select some or all of the data for inclusion in a custom report.

This chapter covers the following topics:

Chapter 7
Geneticist Assistant Reports

Generating a Standard Report

If you want to generate an output file of the data after a run is successfully completed, you can generate a *standard* report. A standard report includes all the data that is displayed in a selected pane on a data tab, for example, the Samples pane on the Run tab. Six different standard reports are available in Geneticist Assistant:

- Run report—You generate the standard Run report from the Run tab. The report includes all the information that is displayed in the Samples pane on the tab.
- Coverage QC report—You generate the Coverage QC report from the Coverage QC tab. The report includes all the information that is displayed on the sub-tabs that you specify—the Coverage Regions sub-tab and/or the Variants sub-tab.
- Panel Statistics reports—You generate a standard Panel Statistics report from the Panel Statistics tab. The report includes all the information that is displayed in any of the following four sub-tabs that you specify—Coverage Regions, Samples, Variants, and/or Patients.
- Sample report—You generate a standard Sample report from the Sample tab. The report includes all the information that is displayed in the panes that you specify— the Variants pane on the Sample Variant Data sub-tab and/or any of the four panes (Status Changes, Patient, Related Patients, or Comments) on the Sample Data sub-tab.
- Variant report—You generate a standard Variant report from the Variant tab. The report includes all the information that is displayed in any of the following five panes that you specify—Variant, Pathogenicity Changes, Artifact Changes, Variant Changes, Variant Comments, Supporting Information, and/or Associated Samples.
- Action List report—You generate the Action List report from the Action List tab. The report includes all the information that is displayed in the panes that you specify—the Variants pane and/or the Coverage Regions pane.

To generate a standard report

1. Open the appropriate tab. See:
   - “The Run Tab” on page 139.
   - “Coverage QC tab (via the Samples pane on the Run tab)” on page 144.
   - “Panel Statistics tab (Run tab)” on page 164.
   - “The Sample Tab” on page 173.
   - “The Variant Tab” on page 190.

To generate an Action List report, you must first create the Action List to open the Action List tab. See "To add selected panel regions or variants to an Action list" on page 162 or "To add selected panel regions or variants to an Action list" on page 182.

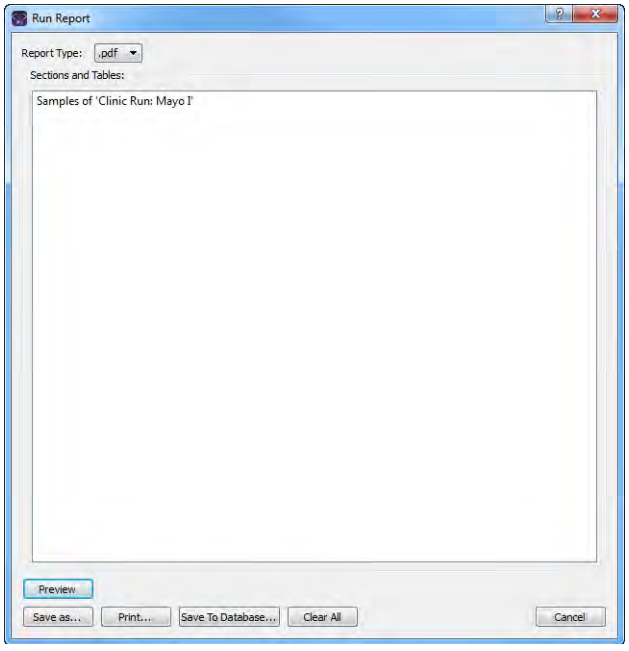
2. Do one of the following:

Table 7-1: Available standard reports

Report to be Generated	Action
Run	On the Geneticist Assistant main menu, click Reports > Run Report.
Coverage QC	On the Geneticist Assistant main menu, click Reports > Coverage QC Report.
Panel Statistics	On the Geneticist Assistant main menu, click Reports > Panel Statistics Report.
Sample	On the Geneticist Assistant main menu, click Reports > Sample Report.
Variant	On the Geneticist Assistant main menu, click Reports > Variant Report.
Action List	On the Geneticist Assistant main menu, click Reports > Action List report.

The appropriate report dialog box, for example, the Run Report or Sample dialog box, opens. All the data that can be selected for inclusion in the report is displayed in the Sections and Tables pane of the dialog box.

Figure 7-1: Run Report dialog box



You can right-click any row and then click "Remove Selected Row" to remove the data from the indicated pane or sub-tab from inclusion in the report.

3. Select the report type.

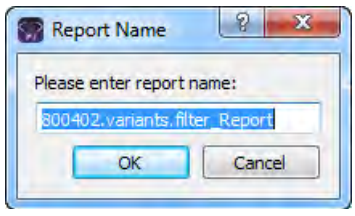
Four formats are available—PDF (the default value), HTML, XML, or CSV, which is a comma-separated values file.

4. Select the options for which to generate the report.

Even if only a single pane is available for report generation (for example, the Samples pane for the Run report), you must still select it. Only the data columns that are shown in the selected pane at the time that the report is generated are included in the report. Hidden data columns are not included. See *"Work with columns in a tab pane" on page 30.*

5. Do any or all of the following as needed:

Table 7-2: Standard report options

Option	Description
Click Preview	To preview the report in the Preview pane on the Report dialog box before you print it and/or save it. After you click Preview, two new options become available: • Close Preview—Close the Preview and leave a blank Report dialog box open. • Refresh—Refresh the preview in real time.
Click Save As	To save the report. You must specify the name of the report and the location for the saved report.
Click Print	To print the report.
Save to Database	To save the report to the Geneticist Assistant database. When you select this option, a Report Name dialog box opens. Regardless of the options that were selected for the report, the same default name. For example, if you generate a standard Sample report, then the default report name is always the sample name. *Figure 7-2: Report Name dialog box*  **Note:** If you have previously saved a report with the default name, then you can enter a new name to distinguish the new report from the previously saved report.

6. Click Close to close the Report dialog box.

Generating a Custom Report

If you want to generate a custom output file of the data after a run is successfully completed, you can generate a *custom* report. A custom report contains only selected data that you specify from a data tab.

To generate a custom report

1. Open the appropriate tab. See:
   - "The Run Tab" on page 139.
   - "Coverage QC tab (via the Samples pane on the Run tab)" on page 144.
   - "Panel Statistics tab (Run tab)" on page 164.
   - "The Sample Tab" on page 173.
   - "The Variant Tab" on page 190.

To generate an Action List report, you must first create the Action List to open the Action List tab. See "To add selected panel regions or variants to an Action list" on page 162 or "To add selected panel regions or variants to an Action list" on page 182.

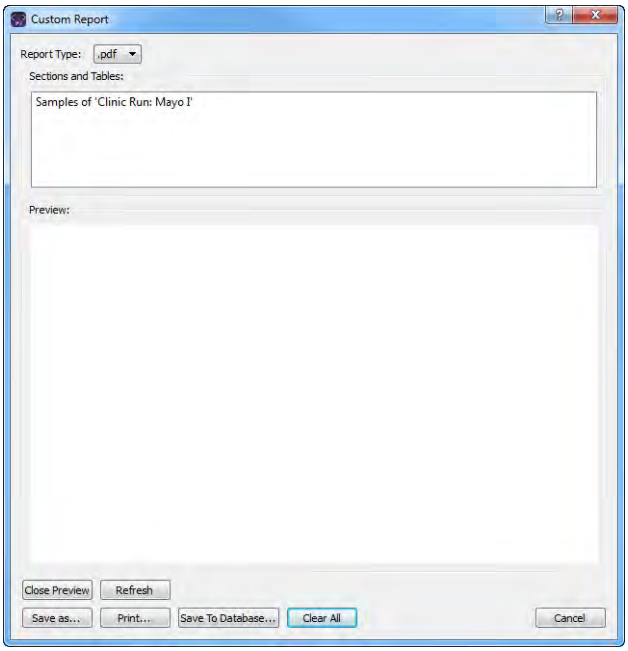
2. Right-click the data that are you including in a custom report, and then select an appropriate option:
   - Add Selected < > to Custom Report—Adds only the data that you select to the custom report. You must press and hold the CTRL key to select multiple items for inclusion in the report.
   - Add All to Custom Report—Adds all the data that is currently displayed in the pane to the custom report.

For both options, only the columns that are shown in the pane can be selected for inclusion in the report. Hidden data columns cannot be selected. See "Work with columns in a tab pane" on page 30.

3. On the Geneticist Assistant main menu, click Reports > Custom Report.

   The Custom Report dialog box opens. All the data that is to be included in the report is displayed in the Sections and Tables pane of the dialog box. See Figure 7-3 on page 210.

Figure 7-3: Custom Report dialog box



4. Optionally, if data has been selected in error for the custom report, or you want to remove some selected data before you generate the report, right-click the entry in the Sections and Tables pane, and on the context menu that opens, click Remove Selected Row.

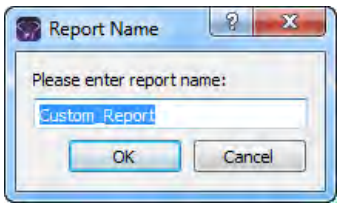
If you previously generated a custom report, and you did not click Clear All before you clicked Cancel, or closed the report dialog box, then any new information that you select on any tab for inclusion in a custom report is simply appended to this existing report. Removing these selected rows is critical to generating the correct report.

5. Select the report type.

Four formats are available—PDF (the default value), HTML, XML, or CSV, which is a comma-separated values file.

6. Do any or all of the following as needed:

Table 7-3: Custom report options

Option	Description
Click Preview	To preview the report in the Preview pane on the Report dialog box before you print it and/or save it. After you click Preview, two new options become available: • Close Preview—Close the Preview and leave a blank Report dialog box open. • Refresh—Refresh the preview in real time.
Click Save As	To save the report. You must specify the name of the report and the location for the saved report.
Click Print	To print the report.
Save to Database	To save the report to the Geneticist Assistant database. When you select this option, a Report Name dialog box opens. Regardless of the options that were selected, the report name is always given the same default name, Custom_Report. *Figure 7-4: Report Name dialog box*  1. Rename the report. 2. Click OK. **Note:** You should rename the report, even though you are not required to do so. Each custom report is saved at its own entry with a unique date and timestamp in the Geneticist Assistant database; however, if you do not change the report name, then each entry is also displayed with the same name in the appropriate Reports pane and therefore, one entry is not distinguishable from another.

7. Click Clear All.

This ensures that information that is selected for a new custom report is not simply appended to an existing report.

8. Click Close to close the Report dialog box.

Chapter 7
Geneticist Assistant Reports

Chapter 8
Geneticist Assistant Tools

Geneticist Assistant contains several tools for working with and analyzing your sample data, including a tool for building a BED file, a tool for managing pathogenicity changes, a tool for specifying pathogenicities for specific variants in a panel, and a tool for combining BAM files into a single sample.

This chapter covers the following topics:

Chapter 8
Geneticist Assistant Tools

Building a BED File with the BED File Builder Tool

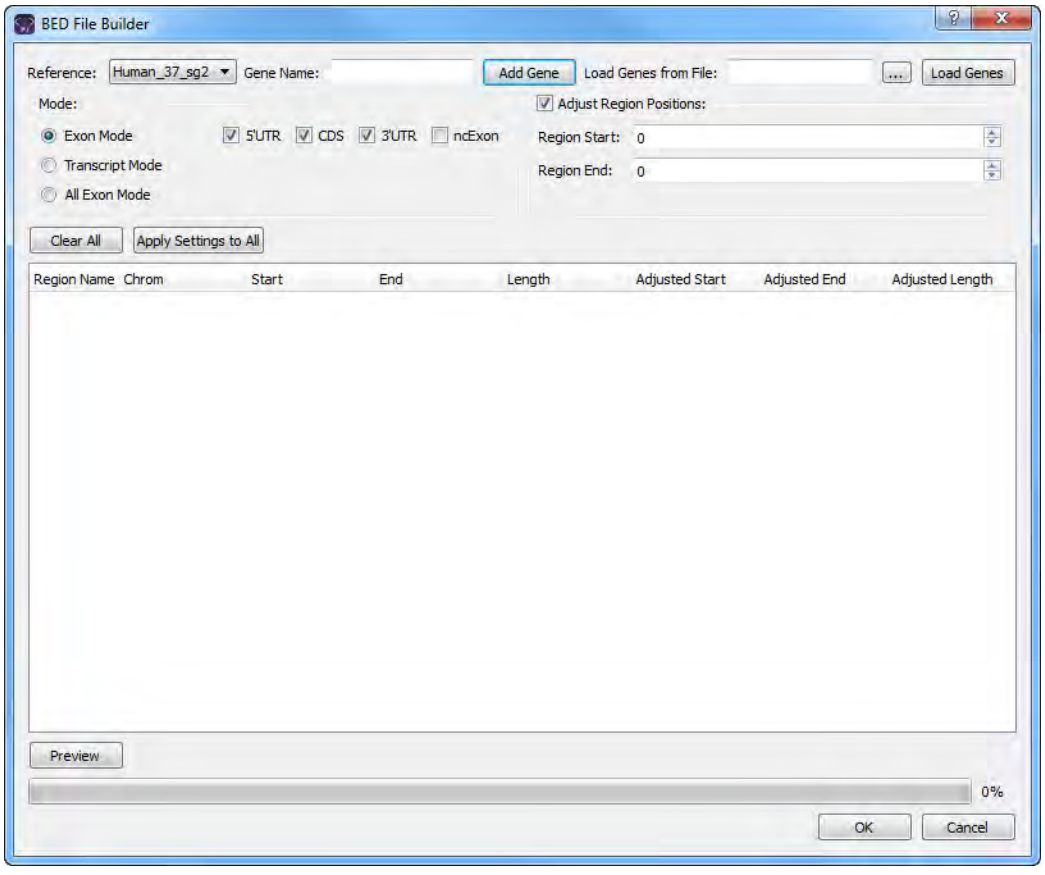
You use BED files to create panels for Geneticist Assistant A BED file defines the genomic regions that are targeted for your particular study or test, which are referred to as *panel regions*. If do not have a BED file for a panel, you can use the BED File Builder tool to build one based on the information that you specify.

To build a BED file with the BED File Builder tool

1. On the Geneticist Assistant main menu, click Tools > BED File Builder.

   The BED File Builder dialog box opens.

Figure 8-1: BED File Builder dialog box



2. Do one of the following:

   - To manually specify the gene name, in the Gene Name field, enter the name for the gene that is in the panel, and then click Add Gene. Repeat this step for each gene that is in the panel.

---

*As you enter the gene name, a list of all genes in the database that meet the search criteria is dynamically updated. You can select the gene from this list. The gene name must begin with the search string, but the search is not case-sensitive. For example, if you enter "**as**" for the search string, then **AS**H1L-AS1 is returned; however, if you enter "**as1**" for the search string, no results are returned.*

   - To load the gene names from a file, click the Browse button ... to open the Select a gene file dialog box, browse to and select the correct file, and then click Load Genes.

The file must be a text file with one line per gene. Optionally, you can specify a transcript for each gene, with a tab between the gene name and the transcript.

   The following information is displayed for all the different transcripts that are available for the gene in the top pane of the dialog box—Region Name, Chromosome, Region Start position, Region End position, and Region Length.

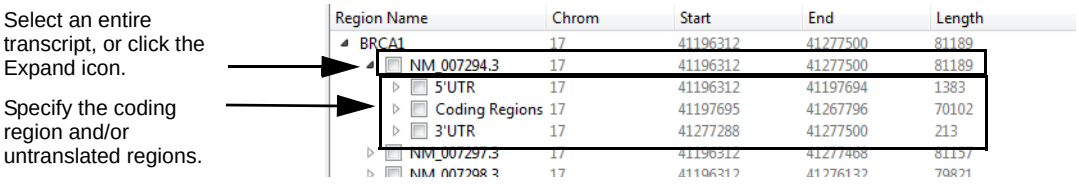
3. Select the mode, which determines the information that is available for creating the BED file.

Mode	Description
Exon	The default mode. You can include only the coding regions in the BED file, only the untranslated regions, or both. Introns are *not* included in Exon mode.
Transcript mode	Adds the full transcript, including introns, to the BED file.
All Exon Mode	Includes all the exons from all the selected transcripts in the BED file.

4. Do one of the following:

   - For Exon mode and All Exon mode—Select the appropriate transcript, or click the Expand icon ▷ next to the transcript, and then select the coding region and/or untranslated regions for building the BED file.

Figure 8-2: BED File Builder tool, Exon mode



   - For Transcript mode—Select the appropriate transcript.

5. Optionally, do any or all of the following as needed:

   - To open the Preview pane and view a preview of the BED file, click Preview.

     The information for each region (Chromosome, Region Start, Region End, Region Name, Score, and Strand) is displayed in the preview.

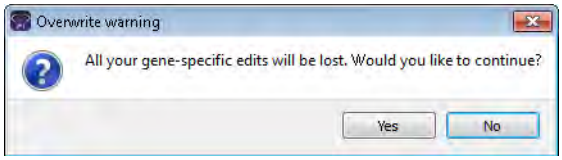
For detailed information about the fields that are displayed in the preview, see https://genome.ucsc.edu/FAQ/FAQformat.html#format1. You can leave the Preview pane open, or you can close it after viewing the BED file. If you leave the pane open, and then make any changes to any of the BED file settings, you must click Refresh to update the preview.

   - To adjust the region start and end positions, enter a different Region Start and/or Region End value (the default value is zero). For example, to include 20 bases at the end of each exon, enter 20 for both the Region Start value and the Region End value.
   - To apply the current settings (the selected mode nd the Start/End Region values) to all genes, including any previously added genes, click Apply to Settings to All.

     An Overwrite warning message opens, indicating that all your gene-specific settings will be lost and asking you if you want to continue.

     Click Yes to close the message and overwrite all the current gene-specific settings with the current gene settings.

Figure 8-3: Overwrite warning message



6. Click OK.

   The Save As dialog box opens. By default, the BED file is named default. bed.

7. Modify the name of the BED file as needed, and save it in a location of your choosing.

Deleting Historical Pathogenicity Changes with the Delete Pathogenicity Changes Tool

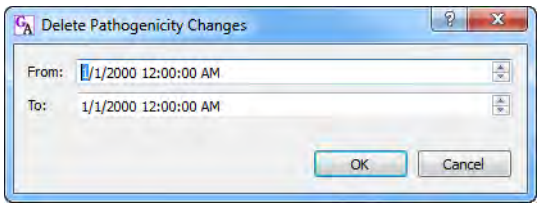
Sometimes, you might determine that a pathogenicity change or changes might not be valid. You use the Delete Pathogenicity Changes Tool to delete all the historical pathogenicity changes for all variants *that were made during a certain time period*. After you delete a historical pathogenicity change for a variant, the pathogenicity of the variant might be reset. For example, if the pathogenicity of a variant was changed from Unknown to Likely Deleterious on January 13, 2013, and you delete all the historical pathogenicity changes that were made from January 1, 2013 to January 31, 2013, then the pathogenicity of the variant is reset to Unassigned. If you changed the pathogenicity of the same variant from Likely Deleterious to Deleterious on February 1, 2013, and you delete all the historical pathogenicity changes that were made from January 1, 2013 to January 31, 2013, then the pathogenicity change from Unassigned to Likely Deleterious is deleted for the variant, but the pathogenicity of the variant remains Deleterious.

To delete historical pathogenicity changes with the Delete Pathogenicity Changes tool

1. On the Geneticist Assistant main menu, click Tools > Delete Pathogenicity Changes.

   The Delete Pathogenicity Changes dialog box opens.

Figure 8-4: Delete Pathogenicity Changes dialog box



2. Change the From value, the To value, or both values as needed. To change the value, select the value that is to be changed, for example, the month, and then you can manually enter the new value, or you can use the Up/Down field arrows to change the value.

You must select each value that is to be changed one at a time. If you need to confirm the time period, remember that the Historical Pathogenicity Changes pane on the Variant tab displays the date and time that a pathogenicity change was made. See "Historical Pathogenicity Changes pane" on page 195.

3. Click OK.

   All the historical pathogenicity changes that were made during the specified time period are deleted from the Geneticist Assistant database. If applicable, the pathogenicities for some variants are reset.

Importing Variants with the Import Variants Tool

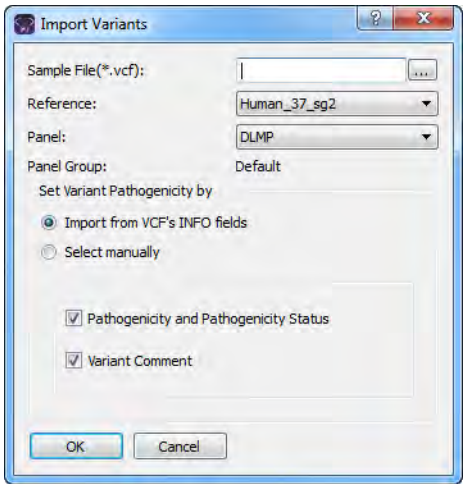
You can use the Import Variant Tool to import all the variants that are contained in a VCF file into your Geneticist Assistant database. If the VCF file contains the pathogenicity and pathogenicity status for the variants and/or comments for the variants, then you can also import this information. If the VCF file's INFO fields do not contain this information, then you can still use the tool to import the variants that are contained in the file, but then you can manually set the pathogenicity and/or pathogenicity status for the variants, as well as add a comment for the variants. For either situation, this means that if multiple variants have the same pathogenicity for a panel group, you do not have to set the pathogenicity of each variant individually. Instead, you can set the same pathogenicity for all the variants at once using this tool.

To import variants with the Import Variants tool

1. On the Geneticist Assistant main menu, click Tools > Import Variants.

   The Import Variants dialog box opens.

Figure 8-5: Import Variants dialog box



2. Next to the Sample File field, click the Browse button ... to open the Select a Variant File dialog box, and then browse to and select the appropriate VCF file.
3. On the Reference dropdown list, select the appropriate reference.
4. On the Panel dropdown list, select the panel for which you are settings the variants' pathogenicity.

   The Panel group name is displayed for Panel Group.

5. Continue to one of the following:
   - "To automatically set the variants' pathogenicities and statuses from the VCF file" below.
   - "To manually set the variants' pathogenicity and status" below.

To automatically set the variants' pathogenicities and statuses from the VCF file

1. Leave Import from VCF's INFO fields selected.
2. Do either or both of the following as needed:
   - If the VCF file contains the pathogenicity and pathogenicity status for each variant, then to import this information, leave the Pathogenicity and Pathogenicity Status option selected. Otherwise, if the VCF file does not contain this information, or you do not want to import this information, then clear this option.
   - If the VCF file contains comments for the variants, then to import this information, leave the Variant Comment option selected. Otherwise, if the VCF file does not contain this information, or you do not want to import this information, then clear this option.
3. Click OK.

   The Import Variants dialog box closes. A Current Jobs tab opens that shows the progress of importing the VCF file. The tab remains open after the import is complete. The variants with the selected information are now imported into the Geneticist Assistant database. These variants will now be displayed with the imported information (such as the pathogenicity) in any samples that contain these variants.

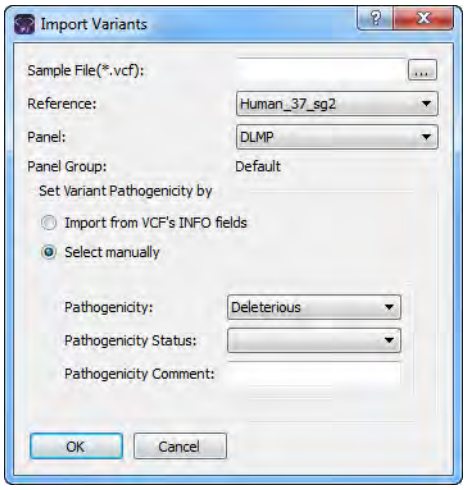
To manually set the variants' pathogenicity and status

When you manually set the pathogenicity and status for the variants in the VCF file, then the pathogenicity and status are set to the *same* values for all the variants in the file. If you add a comment, then the same comment is added for all the variants.

1. Select the Select Manually option.

   The Import Variants dialog box is updated with options for setting the pathogenicity and the pathogenicity status for the variants as well entering a common comment for the variants. See Figure 8-6 on page 221.

Figure 8-6: Import Variants dialog box, Select manually option



2. Do any or all of the following as needed:
   - On the Pathogenicity dropdown list, select the pathogenicity for the variants.
   - On the Pathogenicity Status dropdown list, select the pathogenicity status for the variants.

Remember, you have the option of specifying custom pathogenicity types. Also, by default, only two statuses are available—Pending or Confirmed. You have the option of adding custom pathogenicity statuses. See *"Managing Objects" on page 41.*

   - In the Pathogenicity Comment field, enter a common comment for all the variants.
3. Click OK.

   The Import Variants dialog box closes. A Current Jobs tab opens that shows the progress of importing the VCF file. The tab remains open after the import is complete. The variants with the selected information are now imported into the Geneticist Assistant database. These variants will now be displayed with the specified information (such as the pathogenicity) in any samples that contain these variants.

Merging BAM Files with the Merge BAM Files Tool

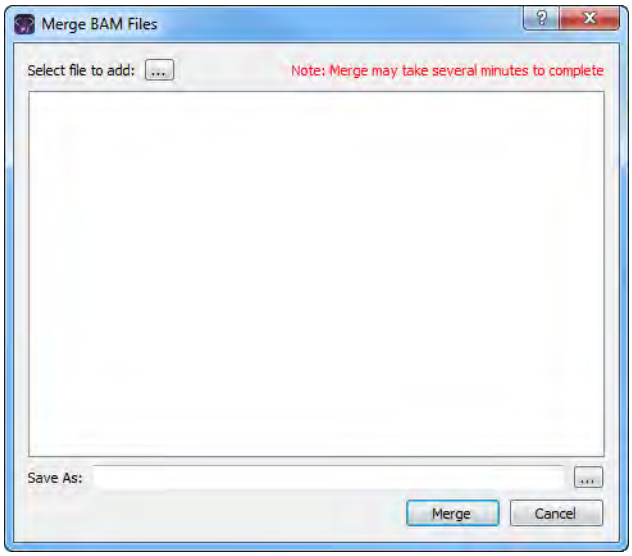
If you have multiple BAM files for a sample, you do not need to submit each file as an individual sample, and then assign each file to the same patient. Instead, you can use the Merge BAM Files tool to merge all the BAM files into one file and then submit the file as a single sample.

To merge BAM files

1. On the Geneticist Assistant main menu, click Tools > Merge BAM Files.

   The Merge BAM Files dialog box opens.

Figure 8-7: Merge BAM Files dialog box



2. At the top of the dialog box, click the Browse button ... to open the Select a file to add dialog box, and browse to and select the BAM files that are to be merged.

Press and hold the CTRL key to select the multiple BAM files.

3. Next to the Save As field, click the Browse button ... to open the Select file to save dialog box, and do the following:
   - In the File name, enter the name for the merged BAM file. (The extension is .bam and you cannot change this.
   - Browse to and select the directory in which to save the merged BAM files.

4. Click Save.

   The Select file to save dialog box closes, and you return to the Merge BAM files dialog box.

5. Click Merge.

   The message “Merging files now” is displayed at the top of the Merge BAM files dialog box. After the BAM files are successfully merged, the message “Merge complete” is displayed at the top of the Merge BAM files dialog box.

Be patient during the merging of the BAM files. Depending on the number of files selected and the sizes of the files, merging might take several minutes.

6. Click Close (x) to close the Merge BAM Files dialog box and return to the Geneticist Assistant main window.

Chapter 8
Geneticist Assistant Tools

Glossary

A

Action List

A list of items that require some type of follow up. In Geneticist Assistant, an Action List can be created for coverage regions on the Coverage QC tab or for variants on the Coverage QC tab or Sample tab.

Administrator user

A user of Geneticist Assistant to whom all available privileges are automatically assigned. Both Administrator privileges and Staff status can be assigned to the same Administrator user.

All Exon Mode

One of the allowed modes for the BED File Builder tool. Includes all the exons from all the selected transcripts in the BED file.

Artifact Type

Indicates that the variant at is not naturally present in the sample, but instead, it is formed by artificial means, such as during preparation of the sample; for example, a Sequencing artifact. A single default value, None, is supplied for artifact type. Additional values can be specified through the Add/Edit Object function in Geneticist Assistant.

Average Coverage

The average number of aligned reads for a region. This value is reported for all panel regions on the Coverage Regions sub-tab on the Coverage QC tab, the Coverage Regions sub-tab on the Panel Statistics tab, and the Coverage Regions tab. A minimum value for the status of a region to be classified as “Passed” can be set on the Quality Control tab of the Settings dialog box.

B

BED file

A BED file is a tab-delimited text file. A panel can be created from a BED file for use in Geneticist Assistant. Each row in the file contains a region of the reference that is to be used in the application. At a minimum, the file contains the following information:

- Field #1 - Chromosome number for the region
- Field #2 - Chromosome start position
- Field #3 - Chromosome end position
- Field #4 - Optional comment column

C

Chromosome Direction

A display setting for sample variants' reference alleles and observed alleles. Display the reference alleles and observed alleles based on the positive strand. Set on the Variant Preferences tab for the Settings dialog box.

Chromosome Position and Annotation pane

Shows the Chromosome Region Start and End positions and annotation for the region that is currently selected on the Coverage Regions sub-tab or for the variant that is currently selected on the Variant sub-tab on the Coverage QC tab.

ClinVar pane

Found on the Variant tab. Displays the information for a variant from the ClinVar database. See http://www.ncbi.nlm.nih.gov/clinvar/intro/.

COSMIC pane

Found on the Variant tab. Displays the information from the COSMIC database for a variant. See http://cancer.sanger.ac.uk/cancergenome/projects/cosmic/about.

Coverage Downstream

The number of base pairs after the end of a panel region to which the quality control settings must apply. The quality control settings are specified on the Quality Control tab of the Settings dialog box.

Coverage Region Status

Indicates whether a panel region passed or failed the coverage settings that were set for the run. The status for every region in a panel is displayed on the Coverage Regions sub-tab on the Coverage QC tab. Default values are Passed or Failed. Additional values can be specified through the Add/Edit Object function in Geneticist Assistant.

Coverage Threshold

The minimum required coverage values that a panel region must meet, or it is flagged as failed region for a run. Thresholds can be set globally (applies to all panel regions in all runs) on the Quality Control tab on the Settings dialog box, or it can set on a per panel basis.

Coverage Upstream

The number of base pairs before the start of a panel region to which the quality control settings must apply. The quality control settings are specified on the Quality Control tab of the Settings dialog box.

Custom report

Contains only selected data as specified by a user.

D

dbNSFP pane

Found on the Variant tab. Displays the information from the dbNSFP database for a variant. See https://sites.google.com/site/jpopgen/dbNSFP.

dbSNP pane

Found on the Variant tab. Displays the information from the dbSNP database for a variant. See http://www.ncbi.nlm.nih.gov/SNP/get_html.cgi?whichHtml=overview.

E

Exome Variant Server pane

Found on the Variant tab. Displays the information from the Exome Variant Server database for the NHLBI GO Exome Sequencing project for a variant. See http://evs.gs.washington.edu/EVS/.

Exon Mode

The default mode for the BED File Builder tool. Allows for the inclusion of the coding regions in the BED file, the untranslated regions, or both.

G

Gene Direction

A display setting for sample variants' reference alleles and observed alleles. Display the reference alleles and observed alleles based on the gene orientation. Set on the Variant Preferences tab for the Settings dialog box.

Group

Two or more users considered a single unit for the purpose of applying the same permissions in a single step.

H

Heterozygosity Lower Bound (%)

The minimum variant frequency for a variant to be considered heterozygous. Set on the Variant Preferences tab of the Settings dialog box. The default value is 20%.

Heterozygosity Upper Bound (%)

The maximum variant frequency for a variant to be considered heterozygous. Set on the Variant Preferences tab of the Settings dialog box. The default value is 80%.

L

LOVD Data pane

Found on the Variant tab. Displays the information from the LOVD (Leiden Open Variation Database) for the variant. See http://www.lovd.nl/3.0/docs/.

M

Minimum Coverage

The minimum coverage depth for a region. This value is reported for all panel regions on the Coverage Regions sub-tab on the Coverage QC tab, the Coverage Regions sub-tab on the Panel Statistics tab, and the Coverage Regions tab. A minimum value for the status of a region to be classified as “Passed” can be set on the Quality Control tab of the Settings dialog box.

Missed Clinical Variant

A clinically relevant position that was identified in the Clinical Variants (ClinVar) database from the NCBI, and that was not covered for the sample.

N

NIST pane

Found on the Variant tab. Displays the information for the variant from the NIST whole human genome Reference Materials database for highly confident variant calls. See http://www.nist.gov/mml/bbd/ppgenomeinabottle2.cfm.

O

Object

Holds a value or variable that can be applied for carrying out runs in Geneticist Assistant.

P

Panel

A collection of genomic regions that are targeted for a specific genomic test or study. Panels are added to Geneticist Assistant by providing a BED file that defines the targeted regions, known as *panel regions*. Each sample that is submitted in Geneticist Assistant is assigned to a specific panel.

Panel Group

Separately tracks the pathogenicities of variants that might be found in multiple panels. For example, the pathogenicity of a selected variant might be set to Deleterious for multiple cancer panels; however, the pathogenicity can be set to benign for a cardiac panel. You can create a group for each panel, and then assign the appropriate panels to each group.

Panel Region

The regions that have been targeted for a specific panel/test as defined by the BED file that was used to create the panel.

Pathogenicity

The pathogenicity for a selected variant, which is the likelihood that the variant will cause a disease. Default values are Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign. Additional values can be specified through the Add/Edit Object function in Geneticist Assistant.

Pathogenicity Status

Indicates the status for changing the pathogenicity for a selected variant; for example, from Unknown to Benign. Default values are Pending and Confirmed. Additional values can be specified through the Add/Edit Object function in Geneticist Assistant.

Patient

Refers to an individual from whom a sample was taken. Any sample that is submitted to Geneticist Assistant can be associated with a patient.

Percent Covered (%)

The percent of a panel region that has reads aligned to it. This value is reported for all panel regions on the Coverage Regions sub-tab on the Coverage QC tab, the Coverage Regions sub-tab on the Panel Statistics tab, and the Coverage Regions tab. A minimum value for the status of a region to be classified as “Passed” can be set on the Quality Control tab of the Settings dialog box.

Preferred Transcript List

A tab-delimited text file that defines the specific transcript or isoform that is to be used for a gene. You can use different transcripts for different panels. The first column in the text file contains the gene name. The second column in the text file specifies the transcript by its nm_accession number.

R

Read Balance

The balance in forward and reverse oriented reads with the variant as reported in the VCF file.

Reference Directory

Contains the information for the reference that was used for aligning data and/or detecting variants.

Run

Refers to loading a BAM or DAT file and a VCF file or just a VCF file into Geneticist Assistant for the purposes of importing a list of variants into the Geneticist Assistant database and/or reviewing a list of variants.

S

Sample Group

Two or more samples considered a single unit for the purpose of repeated processing over a period of time; for example, a group of control samples for which the coverage or variant frequency is to be tracked. Values can be specified through the Add/Edit Object function in Geneticist Assistant.

Sample Group Statistics

Various statistics that can be displayed for a sample group. Displayed on the Sample Variant Data sub-tab for the Sample tab.

Sample Status

The status for a sample. Default values are New, QC Passed, Reviewed, and Complete. Additional values can be specified through the Add/Edit Object function in Geneticist Assistant.

Staff user

A user who can log into the django web administrator site. The actions that the Staff user can carry out depend upon the permissions assigned to him/her. Both Administrator privileges and Staff status can be assigned to the same user.

Standard report

A report that includes all the data that is displayed in a selected pane on a data tab, for example, the Samples pane on the Run tab. Six different standard reports are available in Geneticist Assistant—Run, Coverage QC, Panel Statistics, Sample, Variant, and Action List.

Standard user

A Geneticist Assistant user who has neither Administrator privileges nor Staff status.

Supporting Information

Reference information and/or links to the appropriate websites that have been submitted to support the pathogenicity that is specified for a variant. Displayed in the Supporting Information pane on the Variant tab.

T

Trans

A column on the Variants sub-tab on the Coverage QC tab. Indicates whether the variant is Transition (Ti) or Transversion (Tv).

Transcript mode

One of the allowed modes for the BED File Builder tool. Adds the full transcript, including introns, to the BED file.

U

User

A person who logs into Geneticist Assistant either to add and review content, or use the application in a read-only capacity. Three types of users are available in Geneticist Assistant—an Administrator user, a Staff user, and a standard user.

V

Variant Frequency

The frequency at which a variant is detected in a sample. In Geneticist Assistant, any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.

Variant Preferences

Determines how information about the variants is displayed in all the variant sub-tabs or panes in Geneticist Assistant. Set on the Variant Preferences tab for the Settings dialog box.

Z

Zygosity

Indicates whether the variant is heterozygous or homozygous. In Geneticist Assistant, the selected zygosity is based on the Zygosity settings that are specified on the Variant Preferences tab of the Settings dialog box.

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