GeneticistAssistant™

NGS Interpretative Workbench

User Manual

Customer Support

Customer support is available to organizations that purchase Geneticist Assistant and that have an annual support agreement. Contact SoftGenetics at:

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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 Geneticist Assistant 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 13 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 Geneticist Assistant, including the main menu, the toolbar, the tabs display area, the status bar, and the console.

- Chapter 2, "Geneticist Assistant Setup and Maintenance," on page 53 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, and so on) that you can specify for your Geneticist Assistant installation. It also details quality metrics, which you can use for monitoring and reporting on the NGS data that are you adding to your historical NGS database.

- Chapter 3, "Geneticist Assistant User Management," on page 85 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 and Subpanels," on page 103 details the management of your Geneticist Assistant panels, which includes, adding, editing and deleting panels, and reviewing panel statistics, and adding, editing, and deleting subpanels.

- Chapter 5, "Managing Patients and Runs," on page 125 details the management of patients. It also details how to add and carry out runs in Geneticist Assistant.

- Chapter 6, "Geneticist Assistant Data Review - Runs Tab," on page 177 details all the options that are available to you from the Runs tab for reviewing the data that is generated by a run.

- Chapter 7, "Geneticist Assistant Data Review - Run Tab," on page 187 details all the options that are available to you from the Run tab for reviewing the data that is generated by a run.

- Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209 details all the options that are available to you from the Sample tab for reviewing the data that is generated by a run.

- Chapter 9, "Geneticist Assistant Data Review - Variant Tab," on page 243 details all the options that are available to you from the Variant tab for reviewing the data that is generated by a run.

- Chapter 10, "Geneticist Assistant Reports," on page 265 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. Finally, it details the *Sample PDF report template*, which you can use not only to specify the data that a Sample PDF report is to contain, but also the layout of the report.

- Chapter 11, "Geneticist Assistant Tools," on page 287 details the various tools that are available in Geneticist Assistant to assist you with setting up, running, and analyzing sample data.

- Chapter 12, "The Geneticist Assistant Customer Portal and Submissions," on page 305 details the Geneticist Assistant customer portal, which is a web-based platform that serves as a dedicated hub for tracking and managing your organization's patient and sample information and the tests that have been carried out for each patient/sample combination (a submission). It also describes the Submissions tab, which displays a full record of every submission that any user has ever made through your organization's secure access point to the Geneticist Assistant customer portal.

- Appendix A, "Common Procedures," on page 317 details the procedures that are common to the different Geneticist Assistant entities (run, patient, sample, and variant). Where appropriate, it also details the slight differences that can exist based on the entity for which you are carrying out the procedure.

- The Glossary on page 345 provides detailed definitions of specialized terms for Geneticist Assistant.

Preface

Geneticist Assistant User's Manual

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. Finally, it details how to download and update the necessary reference files, and for data protection, how to review the database log and how to manually back up the Geneticist Assistant database at any time of your choosing.

This chapter covers the following topics:

- "Geneticist Assistant System Recommendations" on page 15.

- "Installing the SoftGenetics License Server" on page 16.

- "Installing Geneticist Assistant on a Windows OS" on page 17.

- "Starting and Logging in to Geneticist Assistant" on page 20.

- "Upgrading Geneticist Assistant" on page 26.

- "The Geneticist Assistant Main Window" on page 27.

- "Setting Display Preferences in the Geneticist Assistant Main Window" on page 31.

- "Searching the Geneticist Assistant Database" on page 46.

- "Updating References" on page 48.

- "Reviewing the Database Log" on page 50.

- "Backing up the Geneticist Assistant Database" on page 51.

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, 8, or 10 64 bit (Recommended) • Windows Vista (64 bit) • Windows Server 2008 (64 bit) • Windows Server 2012 (64 bit)
Processors	Two cores (64 bit)
RAM	• 8 GB (Client) • 4 GB (Server) Minimum: 2 GB for client and server
Hard Drive	• Solid State Drive • 100 GB (Minimum)
Note: Virtual Machines are supported.	

Table 1-2: Geneticist Assistant Data Specifications

Database	Client
• 4 GB disk space (Minimum)	• 150 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 pgdump	
• Low IOPS	

Table 1-3: Geneticist Assistant Software Details

Server	Client	
• Apache httpd 2.4.25 (Latest)	• Qt 5.5.1	• Gsl-lite 0.29.0
• mod_wsgi (Latest)	• Rapidjson 1.1	• Catch
• OpenSSL 1.0.2p (Latest)	• OpenSSL 1.0.2p	• Curl 7.50.0
• Postgres 9.5.14	• LZ4 1.8.0	• Limereport 1.4
• Python 3.6.6 (Latest)	• zlib 1.2.11	
• Django 1.11 (Latest)	• Python 3.6.6	
• South (Latest)	• Htslib	
• Setuptools (Latest)	• Spdlog 0.14.0	
• Virtualenv 16.0.0	• Hayai	

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 in to Geneticist Assistant.

Installing Geneticist Assistant on a Windows OS

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 database server on the same computer or on different computers. The default installation location for both the Geneticist Assistant client and the database server is set to C:\Program Files\Softgenetics\Geneticist Assistant. You can leave the default locations as-is, or you can specify a different location.

Figure 1-1:   Geneticist Assistant Installation wizard, Choose Components page
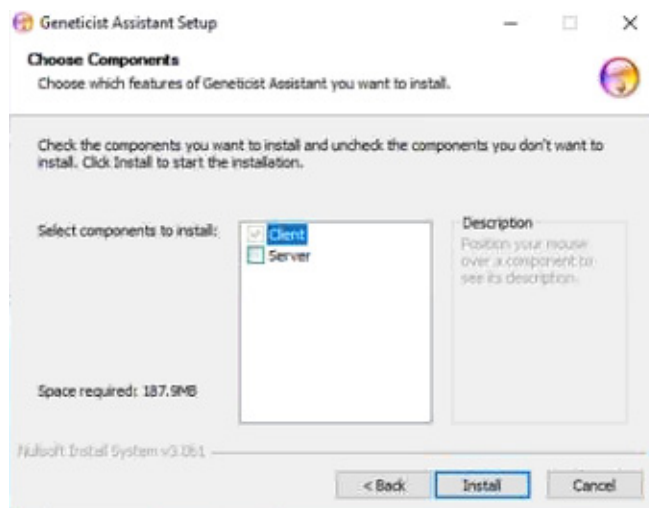
When you are installing Geneticist Assistant, note the following:

- If you are installing the client and database server on the *same* computer, then you must select Server.

- 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.

When you are installing the Geneticist Assistant server, you are prompted to create the Administrator role for the installation and you must configure the server reference directory, which is the database server directory that is to host the Geneticist Assistant reference files. For the Administrator role, 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. The email address is the address that is linked to the Administrator account. See Figure 1-2 and Figure 1-4 on page 18.

Figure 1-2:   Geneticist Assistant Installation wizard, Server Setup, Administrator Configuration page
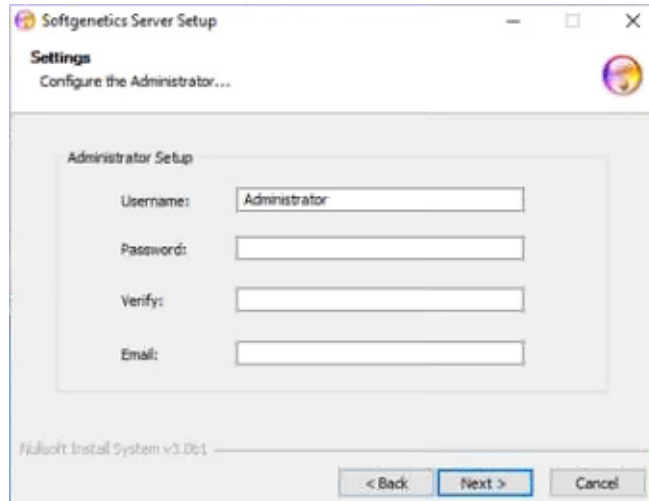
Figure 1-3 below shows the requirements that you can specify for user passwords after you install Geneticist Assistant. For more information about specifying Geneticist Assistant password requirements, see "To specify the Geneticist Assistant password requirements" on page 73.

Figure 1-3:   User password requirement options

Figure 1-4:   Geneticist Assistant Installation wizard, Server Setup, Configure Reference Directory page
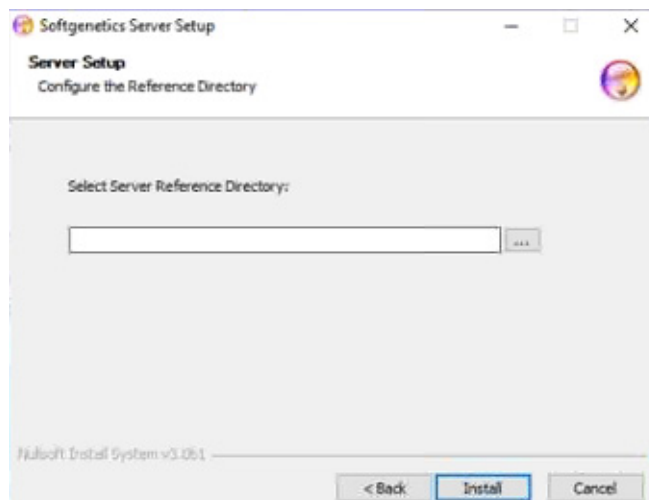
You also have the option of setting the server reference directory when specifying your Geneticist Assistant settings. See "Specifying your Geneticist Assistant Settings" on page 59.

Starting and Logging in to 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.

Figure 1-5:   Geneticist Assistant desktop icon

The results after you start Geneticist Assistant vary, depending on the following:

- Scenario 1 - Whether this is the first time that Geneticist Assistant has been started on the client.

- Scenario 2 - Whether Geneticist Assistant has been started before on the client but the reference directory has not been configured.

- Scenario 3 - Whether Geneticist Assistant has been started before on the client and the reference directory has been configured, but no reference files have been downloaded to the directory.

All the steps in the following procedure are applicable for Scenario 1. The results that you obtain after you start Geneticist Assistant determine where you begin in the procedure for Scenarios 2 and 3. If you have already logged in to Geneticist Assistant, and the reference directory has been configured and reference files have been downloaded to the directory, then continue to "To log in to Geneticist Assistant" on page 22..

To start to Geneticist Assistant

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

   A References Directory Not Found message opens.

Figure 1-6:   References Directory Not Found message
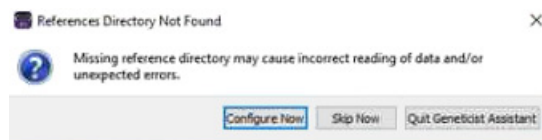
2. Do one of the following:

   - To configure the client reference directory *after* you log in to Geneticist Assistant:

     i   Click Skip.

         The Geneticist Assistant Login dialog box opens.

     ii.   Continue to "To log in to Geneticist Assistant" on page 22.

- To configure the client reference directory *before* you log in to Geneticist Assistant, do the following:

    i   Click Configure Now to open Windows Explorer.

    ii. Browse to and select the appropriate client reference directory.

If the Geneticist Assistant client and server reside on the same computer, then you can set the client reference directory and the server reference directory to the same directory.

    After selecting the directory, a Valid References Not Found message opens.

    iii. Continue to

Figure 1-7:   Valid References Not Found message
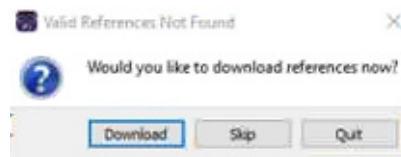
3. Do one of the following:

    - To download the reference files to the client reference directory *after* you log in to Geneticist Assistant:

        i   Click Skip.

        The message closes and the Geneticist Assistant Login dialog box opens.

        ii.   Continue to "To log in to Geneticist Assistant" on page 22.

    - To download the reference files to the client reference directory *before* you log in to Geneticist Assistant:

        i   Click Download.

        The References Downloader dialog box opens.

Figure 1-8:   References Downloader dialog box
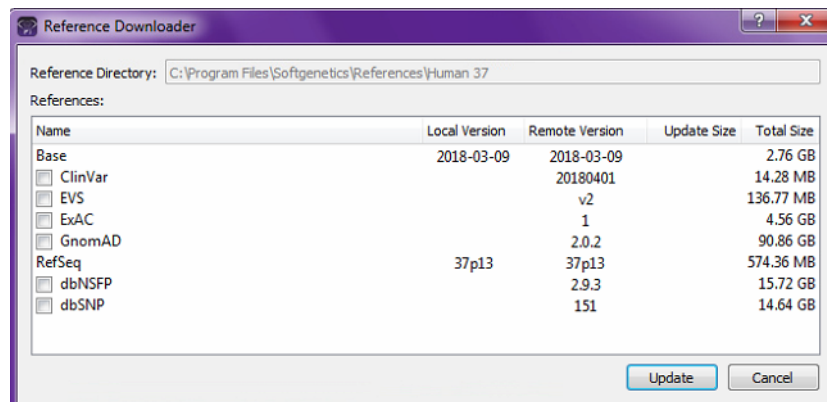
ii.   Select the reference files that you are downloading, and then click Update.

*Base and RefSeq are always selected for download and disabled so that you
cannot clear these options. You can select any or all of the remaining reference
files for downloading/updating. You can also download any of these other
reference files at a later date. See "Updating References" on page 48.*

A Close Geneticist Assistant message opens, indicating that downloading might
require Geneticist Assistant to exit when finished, and asking you if you want to
continue.

Figure 1-9:   Close Geneticist Assistant message
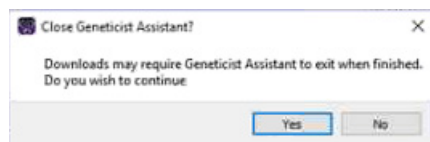
iii. Click Yes.

The message closes and the selected reference files are downloaded to the client
reference directory. If downloading any of these files requires Geneticist Assistant to
close, then after all the selected reference files are downloaded, the Reference
Downloader dialog box closes, and then Geneticist Assistant closes. You must start
and log in to Geneticist Assistant again. See "To log in to Geneticist Assistant"
below.

*Be patient. Depending on the download size, the download process can take up to
several hours.*

To log in to Geneticist Assistant

1.   If you have not already done so, start Geneticist Assistant: Double-click the desktop
icon, or select the option from the Start menu (Start > All Programs > SoftGenetics >
Geneticist Assistant).

The Geneticist Assistant Login dialog box opens. The Server field is automatically
populated with the URL for the database server. The first time that the Login dialog box
opens, the Username field is automatically populated with the user name for the
Administrator user that was set up during installation. For subsequent Geneticist
Assistant sessions, the Username field is automatically populated with the user name for
the user who last logged in to Geneticist Assistant. See Figure 1-10 on page 23.

Figure 1-10:   Geneticist Assistant Login dialog box
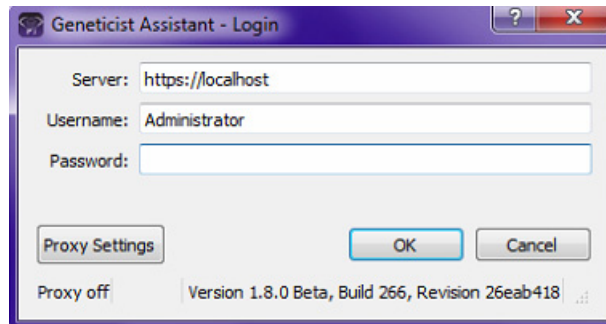
If your organization uses a proxy server for external connections, then to view data in the OMIM and LOVD external databases, you must configure the settings for this proxy server in your Geneticist Assistant instance. Before you log in to Geneticist Assistant, you can click Proxy Settings to open the Proxy pane on the General tab of the Settings dialog box and configure these settings. You can also configure these settings after you log in to Geneticist Assistant. See "To configure proxy server settings" on page 61.

2. In the Username field, enter your username (if applicable), and then in the Password field, enter your password, and then click OK.

3. 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" below.

    •   If you are running a purchased version of Geneticist Assistant, continue to "To run a purchased version of Geneticist Assistant" on page 24.

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 Configure License message opens.

Figure 1-11:   Configure License message
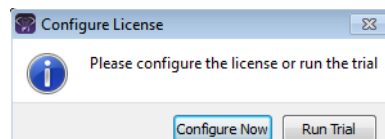
1. Click Run Trial.

    The Configure License message closes.

2. Do one of the following:

    •   If you have already configured the reference directories, then continue in Geneticist Assistant as appropriate.

- If you have yet to configure the reference directories, then before you can continue in Geneticist Assistant, at a minimum, you must create your server and client reference directories, specify their locations, and then download reference files to the client reference directory. You must also specify the global Quality Control (coverage) settings for your data. See "Specifying your Geneticist Assistant Settings" on page 59.

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 Geneticist Assistant, a Configure License 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 1-12:   Configure License message
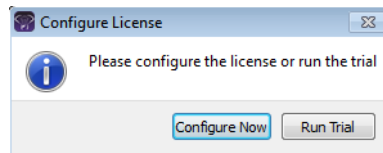
1. Click Configure Now.

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

Figure 1-13:   Settings dialog box
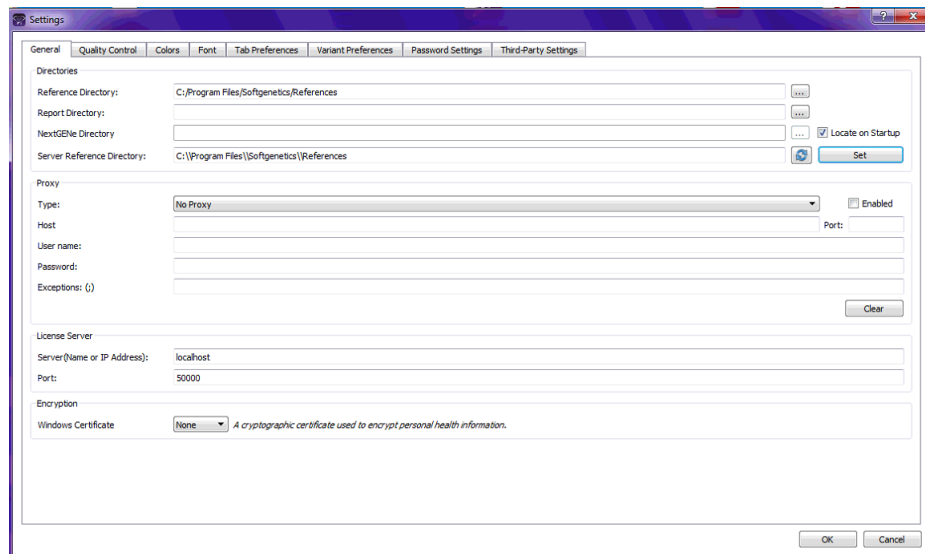
2. In the Server field, enter the fully qualified domain name (FQDN) for the server (where the license server manager is installed), or its IP address.

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

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

4. Click OK.

    The Settings dialog box closes.

5. Do one of the following:

    •   If you have already configured the reference directories, then continue in Geneticist Assistant as appropriate.

    •   If you have yet to configure the server reference directory, then before you can continue in Geneticist Assistant, at a minimum, you must create your server and client reference directories, specify their locations, and then download reference files to the client reference directory. You must also specify the global Quality Control (coverage) settings for your data. See "Specifying your Geneticist Assistant Settings" on page 59.

To log out of Geneticist Assistant

To log out of Geneticist Assistant, on the main menu, click File > Exit.

Upgrading Geneticist Assistant

If you are upgrading to a newer version of Geneticist Assistant, then you do not need to uninstall the current version. If you are upgrading from any version prior to Version 1.4, then after you have completed the upgrade, and log in to Geneticist Assistant for the first time, an Uncompressed References Detected message opens. The messages states that Geneticist Assistant first needs to compress all the associated reference files before you can work in Geneticist Assistant.

Figure 1-14:   Uncompressed References Detected message dialog box
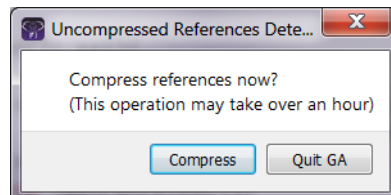
1. Click Compress.

   A dialog box opens, indicating the progress in compressing the reference files.

Figure 1-15:   Compressing reference files progress dialog box
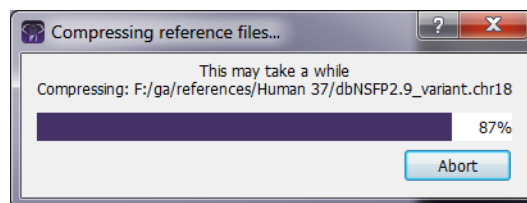
This process can take anywhere from one to four hours based on your system's available resources, and Geneticist Assistant is unavailable during this time.

2. After the Compression Progress dialog box indicates that the compression is 100% complete, click OK.

   The Compression dialog box closes, and Geneticist Assistant is now ready for use.

The Geneticist Assistant Main Window

The Geneticist Assistant main window opens automatically after you successfully log in to Geneticist Assistant.

Figure 1-16:   Geneticist Assistant main window
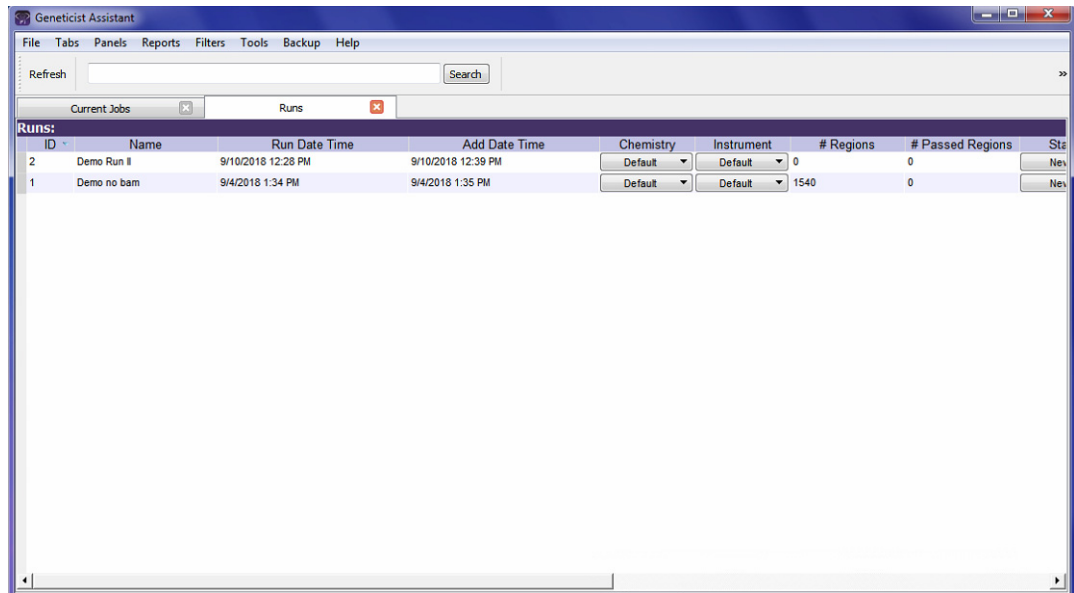
The Geneticist Assistant main window is your starting point for Geneticist Assistant. The window provides quick access to all of the Geneticist Assistant functions. From top to bottom, the Geneticist Assistant main window has five major permanent components—the title bar, the main menu, the toolbar, the tabs display area, and the status bar—and one transient component, the console.

Title bar

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

Figure 1-17:   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, Tabs, Reports, Filters, Tools, Backup, and Help) across the menu bar.

Figure 1-18:   Geneticist Assistant main menu
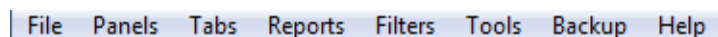
Toolbar

The Geneticist Assistant main window toolbar is located below the main menu. It always contains a Refresh option, a Search feature, and Ref option. 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, editing, 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, or variants in your Geneticist Assistant database. See "Searching the Geneticist Assistant Database" on page 46.

- You use the Ref option to update any reference that has been downloaded to the Geneticist Assistant database to the most current version. See "Updating References" on page 48.

Figure 1-19:   Geneticist Assistant main window toolbar
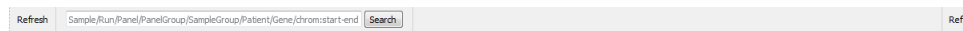
Figure 1-20:   Geneticist Assistant main window toolbar for the Runs tab
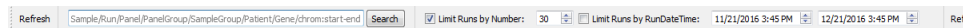
Main window tabs

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 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 1-21:   Geneticist Assistant main window, Runs tab with Runs pane
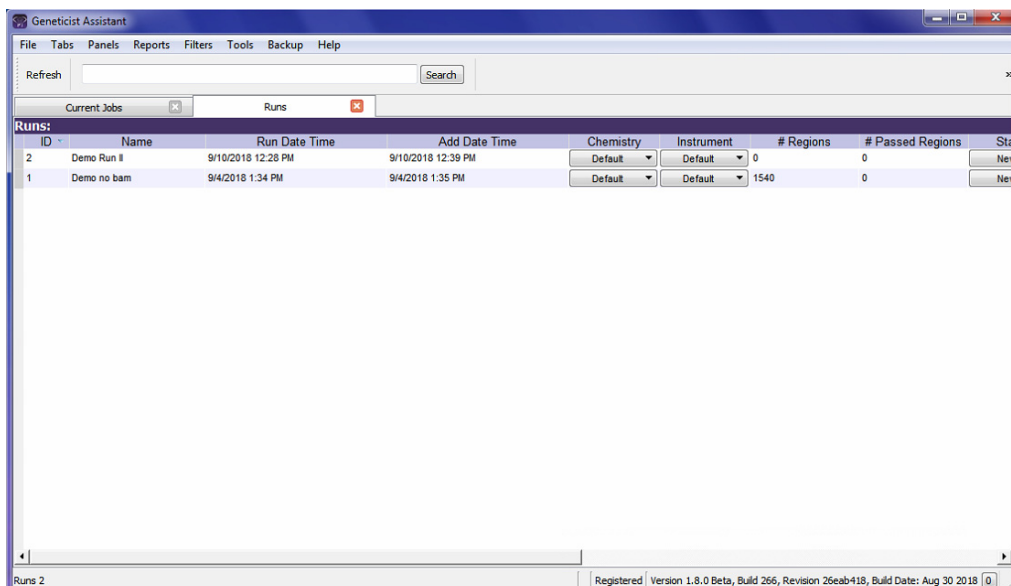
- If you are an Administrator user, then the Runs pane can display all the runs submitted by all users to your Geneticist Assistant database. By default, when the tab first opens, only the 30 most recently submitted runs are displayed and the runs are sorted based on the Run ID. You can limit the runs 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 the 30 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, or any other process that is currently being carried out in Geneticist Assistant. The tab opens automatically after a run is started. After the run is successfully completed (a BAM 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 1-22:   Geneticist Assistant main window, Status bar

| Runs 13 | | Registered | Version 1.8.0 Beta, Build 1, Revision e8219bb5, Build Date: Oct 18 2018 | 0 |
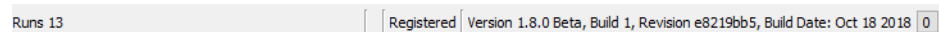
Console

The console is displayed below the status bar at the bottom of the Geneticist Assistant main window. The console is a transient component of the main window and it is automatically opened and displayed whenever an error occurs in the software, for example., a required file is not specified. You can click the Close ("x") button in the upper right corner of the console

to close it. The console remains closed until another error occurs, and then it again opens automatically and displays the error.

Figure 1-23:   Geneticist Assistant main window, console

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 "To navigate multiple open tabs/ Navigate tabs with a minimized main window" below.

- Close multiple tabs in a single step. See "To close multiple tabs in a single step" on page 32.

- Filter the data that is displayed in a tab pane. See "To filter the data that is displayed in a tab pane" on page 32.

- Show/hide data columns in a tab pane. See "To show/hide columns in a tab pane" on page 39.

- Manage tab pane configurations. See "To manage tab pane configurations" on page 41.

- Change the widths of a data columns. See "To change the widths of data columns" on page 44.

- Rearrange the order of data columns. See "To rearrange the order of data columns" on page 44.

- Change the sort order of data. See "To change the sort order of data" on page 44.

- Rename column headers. See "To rename column headers" on page 45.

- Select the data that is to be included in a custom report. See "To select data to be included in a custom report" on page 45.

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 any Variants pane or tab, for example, the Sample Variant Data sub-tab on the Sample tab, and then apply these same settings to all the other Variants panes and tabs in Geneticist Assistant. 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 69.

To 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. 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.

To 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.

To filter the data that is displayed in a tab pane

By default, when a tab first opens, all data in all available panes is displayed. You can open a Table Filters pane for a selected tab pane which displays all the data (fields) for the pane, and then select from the fields that are listed to filter the pane display 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 template, and then load the template to filter the data that is displayed in the tab pane anytime as needed. You can also delete a saved filter template.

1. Open the appropriate tab.

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

   The Table Filters pane opens to the right of the tab. The pane is docked to the right side of the Geneticist Assistant main window. The pane consists of two sub-panes. The left sub-pane displays all the fields for the selected tab pane. The right sub-pane is blank.

3. Repeat Step 2 as many times as needed to open multiple Table Filters panes for the tab.

Figure 1-24:   Multiple Table Filters panes opened for a tab
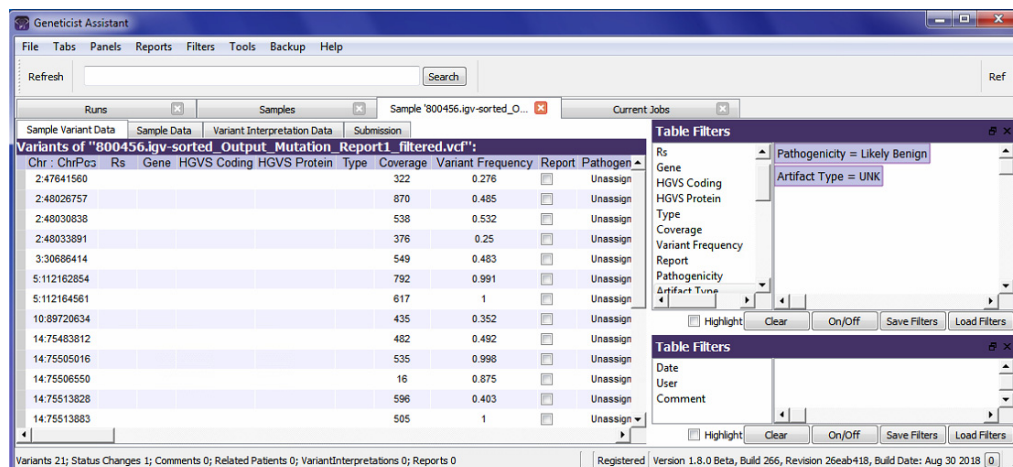
The Table Filters panes are docked from top to bottom in the order in which you opened them. Optionally, to make a Table Filters pane a floating pane, you can click the Undock icon that is in the top right corner of the pane (the icon that is to the left of the Close (x) icon), and then drag the pane. A floating pane is a pane that is visible in the Geneticist Assistant main window, but its location is not confined to a specific area of the window. Instead, you can independently change its size and/or position, including moving the pane outside the main window.To redock the Table Filters pane, click the Close (x) icon to close the pane, and then on the Geneticist Assistant main menu, click Filters and select the tab pane again.

The remainder of this procedure describes how to set up new a data filter. Optionally, below a Table Filters pane, you can click Load Filters to open the Load Filter Template dialog box and select from a list of saved filter templates for the pane.

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 Edit Filter dialog box displays different options depending on the selected filter.

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 1-25: Edit Filter dialog box example - Zygosity column selected
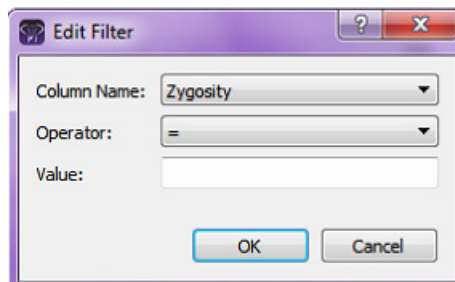
For example:

*   If you are filtering data based on a date/time field, then a Relative option is displayed on the Edit Filter dialog box. You select this option to display data from a specified time period that is relative to the current date and time. For example, to display data that has been generated within the last 30 days of the current date and time, select the < operator, select Relative, and then in the Relative field, enter -30 days. (You must enter the minus sign.) See Figure 1-26 on page 34.

Figure 1-26:   Edit Filter dialog box, Date/Time selected as filter
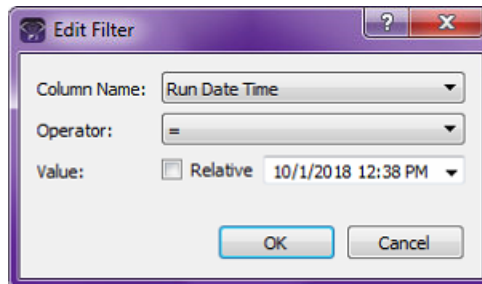
•   If you select to filter by Gene, then a Load by phenotype option is displayed on the Edit Filter dialog box. Enter a phenotype to filter by genes that are associated with this phenotype. As you enter the search string, a dropdown list opens. The list is dynamically updated with the phenotypes that are found in the Human Phenotype Ontology database that match the search string.

*The search string is not case-sensitive, and the string can be found anywhere in the search results. For example, a search string of "thyroid" returns phenotypes of **thyroid** adenoma and secondary hyper**thyroid**oism.*

Figure 1-27:   Edit Filter dialog box, Gene selected as filter
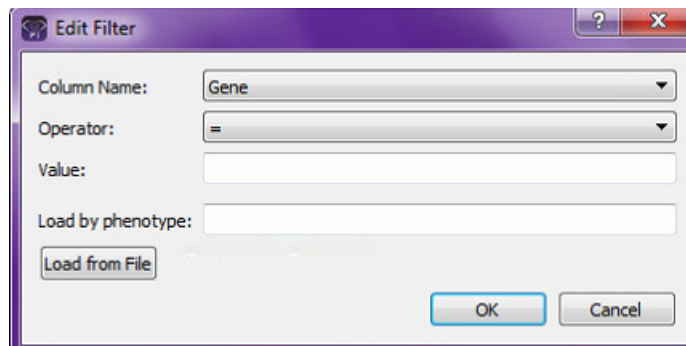
5.   On the Operator dropdown list, select the operator for the filter.

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.
≠	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.

Filter Operator	Description
>=	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. In the Edit Filter dialog box, enter the values for the filter. The dialog box displays different options depending on the selected filter. For example:

   - You might have only the option of entering a value in the Value field. If you enter multiple values, then you use a comma to separate multiple values.

   - You might have the option of entering a value in the Value field, or clicking a Load Values option to open the Open Text or CSV file dialog box, and then browsing to and selecting the appropriate file.

   A text file contains a single value. A Comma-Separated Values (CSV) file contains multiple values that are separated by commas.

7. Click OK.

   The Edit Filter dialog box closes. The selected field, its operator, and its value are displayed in the right sub-pane. The filter is applied immediately to the pane.

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 additional fields for filtering *on top* of an existing filter field to apply the filters on an "and" basis. For example, for the Sample Variant Data 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 that is greater than or equal to 300 *and* that are

found in the MSH6 gene.

- You can drag separate additional fields for filtering into the blank pane to apply filters on an "or" basis. For example, for the Sample Variant Data 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 that is greater than or equal to 300 *or* variants that are found in the MSH6 gene.

Figure 1-28:   And and Or filters
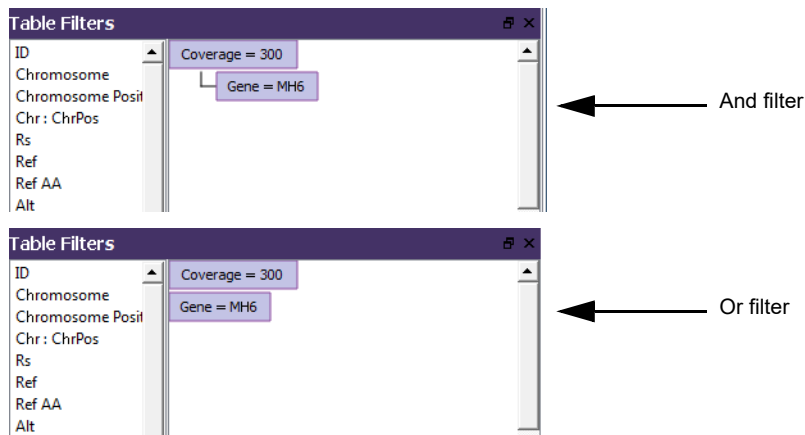
9. Do any of the following as needed for a specific Table Filter:

- To highlight the results in bright yellow that pass the filtering criteria instead of removing data that fails the filtering criteria, select Highlight.

- The On/Off option is a toggle.

  - To remove the applied filters, click On/Off.

  - To apply all the filters and view the results onscreen, click On/Off again. ("*Filters Applied" is displayed in the title bar of the affected tab pane.)

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

- To remove all the filters that have been applied for the tab pane, click Clear. The filters remain displayed in the Table Filters pane.

To clear all filters for all tabs in a single step, on the Geneticist Assistant main menu, click Filters > Clear All Filters.

- To edit a filter, right-click the filter and on the context menu that opens, click Edit to open the Edit Filter dialog box. Edit the filter (Column, Operator, and/or Value) as needed, and then click OK to close the dialog box and immediately save and apply the edited filter.

- To delete a filter, right-click the filter and on the context menu that opens, click

Remove. The pane display is immediately updated.

- To save your filter settings, click Save Filters to open the Save Filter Template dialog box and then do the following:

Figure 1-29: Save Filter Template dialog box
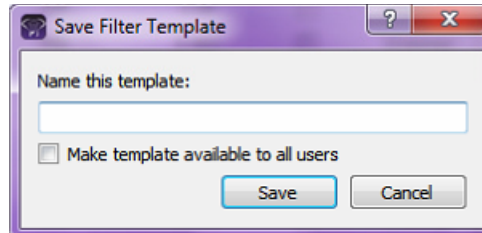
i   Enter a name for the filter template.

ii.   If this template is to be available to other users, then select Make template available to other users; otherwise, the template is available only to you.

iii.   Click Save.

A Template Save Result message opens, indicating that the new template was saved.

Figure 1-30: Template Save Result
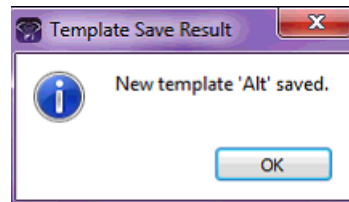
iv.   Click OK to close the message and save the filter template to the Geneticist Assistant database.

You, and if applicable, other users, can use this saved filter template to filter the data for this specific tab pane for another project based on the template settings.

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

To load a saved filter template

You can load any filter template that you have added to the Geneticist Assistant database and you can open any filter template that another user has added and made available to all users.

1. Open the appropriate tab.

2. On the Geneticist Assistant main menu, click Filters > <Tab Pane>. For example, if you clicked 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 Table Filters pane opens to the right of the tab. See

3. Click Load Filters.

   The Load Filter Template dialog box opens. The dialog box displays all the filter templates that have been saved and made available to all users for the selected tab pane.

Figure 1-31:   Load Filter Template dialog box

4. Select the filter template that you are loading, and then click Load.

   The Load Filter Template dialog box closes and the selected filter is applied immediately. The data in the tab pane is updated to reflect the filtering.

To delete a saved filter template

You can delete filter templates that have been saved by any user for any tab pane.
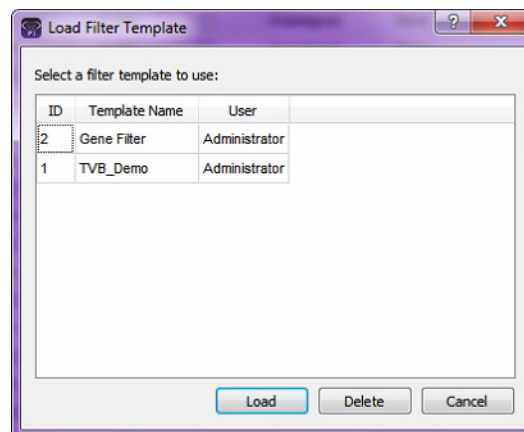
1. Open the appropriate tab.

2. On the Geneticist Assistant main menu, click Filters > <Tab Pane>. For example, if you clicked 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 Table Filters pane opens to the right of the tab. See

3. Click Load Filters.

The Load Filter Template dialog box opens. The dialog box displays all the filter templates that have been saved by all users for the selected tab pane.

Figure 1-32:   Load Filter Template dialog box
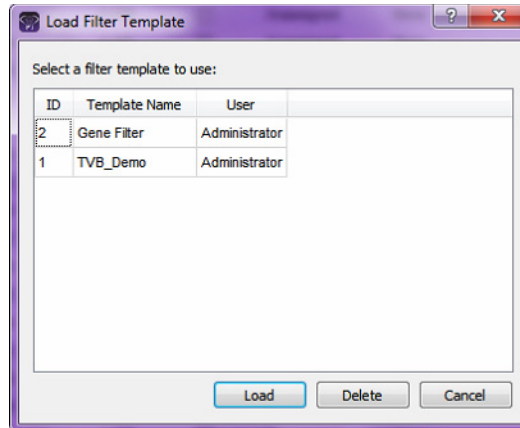
4. Select the filter template that you are deleting, and then click Delete.

The filter is deleted immediately. The Load Filter Template dialog box remains open.

To show/hide columns in a tab pane

You specify the columns that you are showing or hiding in a tab pane in the Select Columns dialog box. By default, when the dialog box first opens for a tab pane, the columns that are currently displayed are selected, and Select/deselect/restore all is either selected or completely filled. The Select/deselect/restore all option on the Select Columns dialog box is a tri-state checkbox. When the dialog box is opened for the first time for a tab pane, all the columns are selected for display, and the Select/deselect/restore option is also selected. After you make your column selections, then each subsequent time that the dialog box opens, the columns that have been selected for display are selected, those that have been hidden are deselected, and the Select/deselect/restore option is completely filled. If you click this option once, then all columns are shown in the tab pane in a single step and the select/deselect/ restore option is selected. If you click the Select/deselect/restore option a second time, then all columns are hidden in the tab pane in a single step, and the option is cleared. If you click the Select/deselect/restore option a third time, then the last specified settings are restored, and a completely filled checkbox is displayed for the option.

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

A sub-menu opens that displays up to three options: Manually, Compact, and all the available configurations for the pane. The name of the user who created a configuration is displayed next to the configuration.
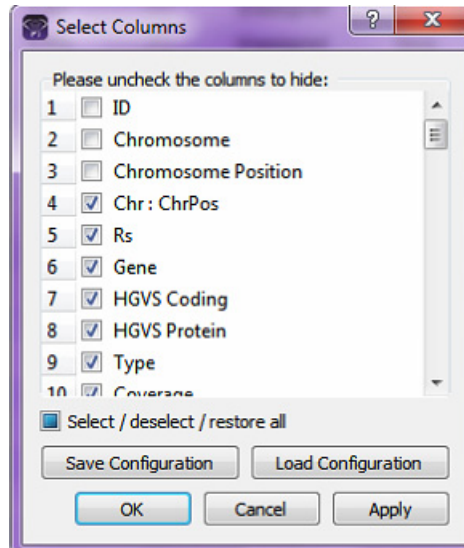
Compact displays the data columns in the pane in a more condensed view. To toggle the display, click Compact as needed.

2. Click Manually.

    The Select Columns dialog box opens.

Figure 1-33:   Select Columns dialog box

3. If a column is to be hidden from view in the pane, then clear its selection; otherwise, leave it selected.

4. Optionally, to rearrange the column display, click the column number for a column, and then drag it to a new location in the list of columns.

This is identical to rearranging the data columns directly in a tab pane. See "To rearrange the order of data columns" on page 44.

5. Click Apply to update the column display.

6. Click OK 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 manage tab pane configurations

Instead of selecting individual columns to show or hide, you can use pane configurations to manage your data column displays. You can load an existing pane configuration, you can create your own pane configurations, and you can delete pane configurations. See:

- "To load a pane configuration" below.

- "To create/edit a pane configuration" below.

- "To delete a pane configuration" on page 43.

To load a pane configuration

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

    A sub-menu opens that displays up to three options: Manually, Compact, and all the available configurations for the pane. The name of the user who created each configuration displayed next to the configuration.

    Compact displays the data columns in the pane in a more condensed view. To toggle the display, click Compact as needed.

2. Select the configuration that you are loading.

    The context menu closes. The pane display is updated dynamically after the configuration is loaded.

To create/edit 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 edit only the configurations that you have created.
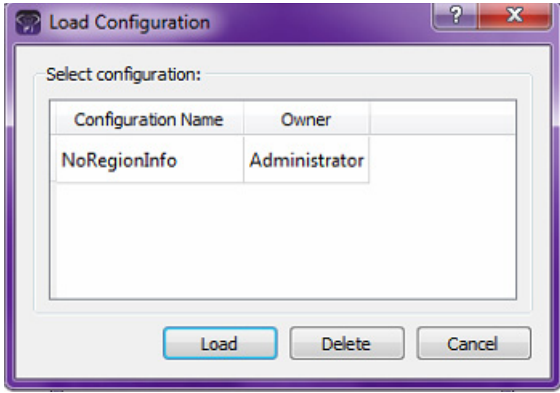
1. Right-click the tab pane anywhere, including the column headers, and on the context menu that opens, click Load Columns.

    A sub-menu opens that displays up to three options: Manually, Compact, and all the available configurations for the pane. The name of the user who created each configuration displayed next to the configuration.

    Compact displays the data columns in the pane in a more condensed view. To toggle the display, click Compact as needed.

2. Do one of the following:

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, the columns that are currently displayed are selected, and Select/deselect/restore is either selected or completely filled. See Figure 1-33 on page 40. 2. Continue to Step 3.
To create a new pane configuration by editing an existing configuration	1. Click Manually. The Select Columns dialog box opens. See Figure 1-33 on page 40. 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 1-34: 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.

3. Clear the options for the columns that you are hiding from the pane view, and select the options for the columns that you are displaying in the pane view.

4. Click Apply to update the column display.

5. Click Save Configuration.

The Save Configuration dialog box opens.

Figure 1-35: Save Configuration dialog box
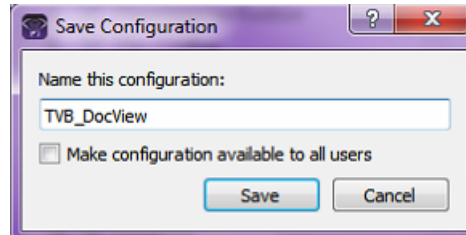
6. Enter a name for the configuration.

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

7. If this configuration is to be available to all other users 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.

8. Click Save.

The Save Configuration dialog box closes. The Select Columns dialog box remains open.

9. Click OK to close the Select Columns dialog box.

To delete a pane configuration

You can delete only those pane configurations that you have created, and you cannot delete the default System configuration.

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

A sub-menu opens that displays up to three options: Manually, Compact, and all the available configurations for the pane. The name of the user who created each configuration displayed next to the configuration.
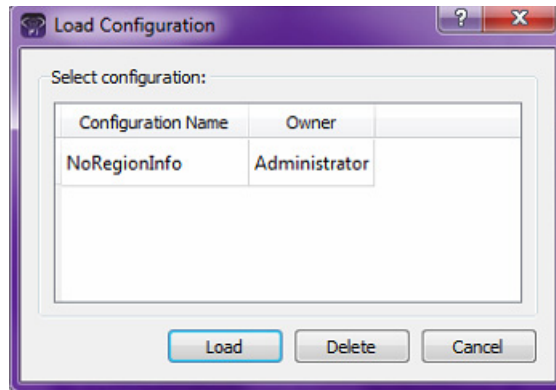
2. Click Manually.

The Select Columns dialog box opens. See

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. See

Figure 1-36:   Load Configuration dialog box

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 OK to close the Select Columns dialog box.

To 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.

To 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.

You can also rearrange the order of data columns in a tab pane using the function that is available in the Select Columns dialog box. See "To show/hide columns in a tab pane" on page 39.

To 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.

To 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 edit the column header as needed.

To 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 (CTRL-click 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 271.

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 1-37: Geneticist Assistant main window toolbar, Search feature
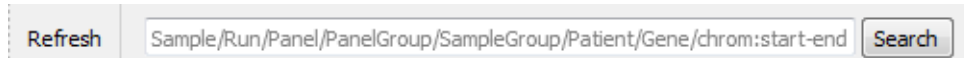
Important points to note about a search are the following:

- 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).

- With the exception of searching by HGVS, 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."


Searching by HGVS supports only a perfect match to the search string, for example, p.Asn1983Ser.

- 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.

  - You can search by HGVS variant nomenclature.

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

Figure 1-38: Search feature and results example
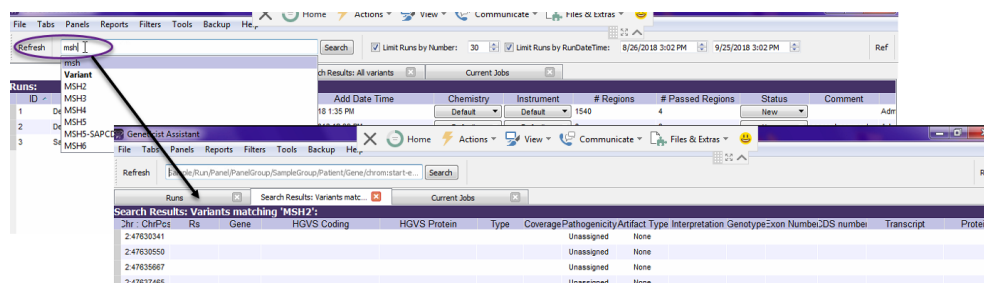
You can double-click any search result on a Search Results tab to "drill down" into the search result, and review the associated data. For example, you can:

- 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.

Updating References

The *reference directory* contains the information for the reference that was used for aligning the data and/or detecting variants in sample. References are stored in their own folders in the references directory. SoftGenetics initially provides the necessary references files for your Geneticist Assistant installation. You use the Ref function that is available on the toolbar of the Geneticist Assistant main window to update to the most recent version of any reference in the reference directory. You can select multiple references for updating at the same time.

## To update a reference

1. On the toolbar of the Geneticist Assistant main window, click Ref.

   The Reference Downloader dialog box opens. The dialog box lists the following information for each reference that is currently installed for your Geneticist Assistant instance:

   - The reference name.

   - The Local Version, which is the version of the currently installed reference.

   - The Remote Version, which is the most recent version that is available on the SoftGenetics server.

   - The Update Size, which is the difference in size between the Local Version and the Remote Version.

   - The Total Size of the downloaded reference.

   The location of the Reference Directory that has been specified for your Geneticist Assistant instance is displayed at the top of the dialog box. A checkbox is displayed next to each reference for which the Local Version and Remote Version are different.

   Figure 1-39: References Downloader dialog box
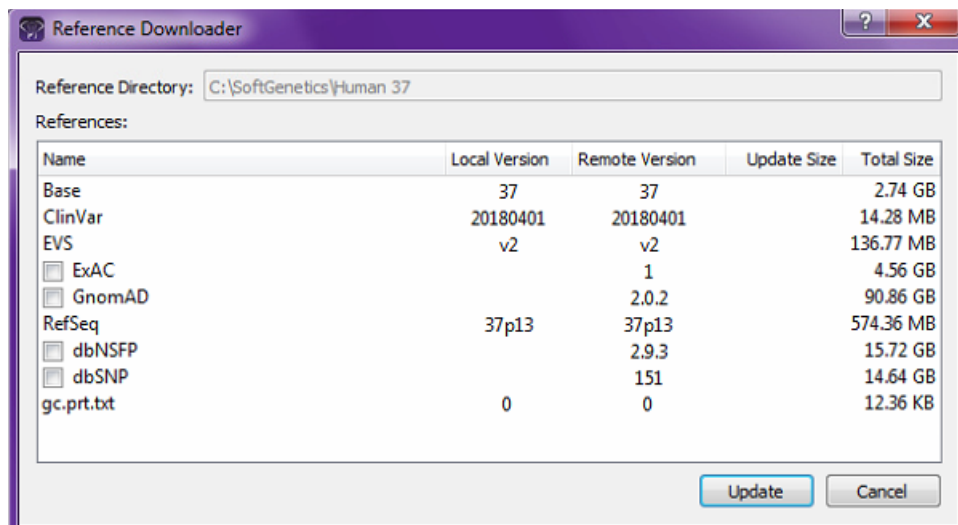
The first time that you download reference files, Base and RefSeq are always selected for download and disabled so that you cannot clear these options. You can select any or all the remaining reference files for downloading/updating. You can also download any of these other reference files at a later date. When you are updating references, because Base and RefSeq have already been downloaded, they are not selected and disabled.

2. Select the references that you are updating, and then click Update.

   After the selected references are updated, the Local Version and the Remote Version are identical.

Be patient. Depending on the update size, the update process can take up to several hours.

Reviewing the Database Log

Geneticist Assistant tracks *all* actions that have occurred for its database, such as user logins, data additions, data edits, data deletions, and so on, in a database log. This database log is displayed on the Database Log tab. The log shows all the actions that have occurred in the Geneticist Assistant database in reverse chronological order. For each action, the following information is displayed: the date/time of the database action, the user who carried out the action, the type of action, and the data type that was affected by the action. A detailed description of the action is also supplied. To review the database log, on the Geneticist Assistant main menu, click File > Database Log.

Figure 1-40:   Database Log tab
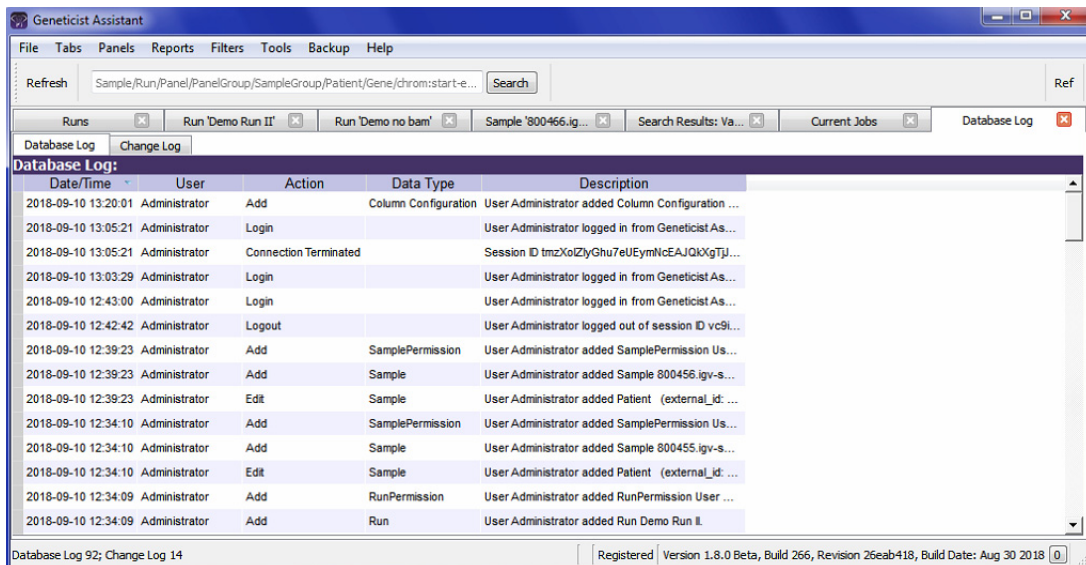
Backing up the Geneticist Assistant Database

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 > Database Backup.

   The Database Backup dialog box opens. In the Output field, the default name and location for saving the database backup file is set to C:\Program Files\SoftGenetics\Geneticist Assistant\ga_exe\softgenetics_backup_<current year>_<current month_day>_<current_time>. You have the option of changing one or both of these default values.

Figure 1-41:   Database Backup dialog box
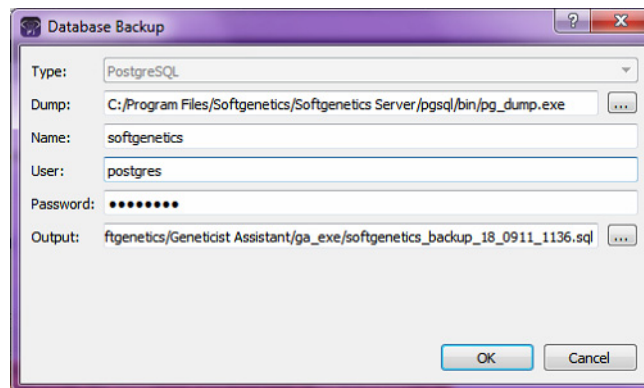
2. Next to the Dump path field, click the Browse button […] to open the Select dump tool dialog box, and then browse to and select pg_dump.exe file.

   The default installation directory for the Postgres dump tool is C:/Program Files/ Softgenetics/Softgenetics Server/pgsql/bin/pg_dump.exe.

3. Optionally, do one or both of the following:

   • To change the name of the database backup file, in the Name field, enter the new value.

     For example, to change the name from "softgenetics_backup_<current year>_<current month_day>_<current_time>" to "geneticist_assistant_backup_<current year>_<current month_day>_<current_time>", enter geneticist_assistant in the Name field.

   • To change the location for the database backup file, next to the Output field, click the Browse button […] to open the Select Output directory dialog box, and then select a

different location for the database backup file.

4. Enter your Postgres username and password.

Make sure to enter your Postgres login credentials, and NOT your Geneticist Assistant login credentials. Unless you have customized your Postgres installation, the username is postgres and the password is softgeneticssetup.

5. Click OK.

After the Geneticist Assistant database is successfully backed up, the Database Backup dialog box closes. The Geneticist Assistant main window remains open.

Chapter 2

Geneticist Assistant Setup and Maintenance

Before you use Geneticist Assistant for the first time, you must add and/or edit the necessary objects and specify the settings for your installation. Optionally, you can specify quality metrics for monitoring runs and ensuring that the runs results are meeting your organization's performance standards.

This chapter covers the following topics:

*   "Managing Objects in the Geneticist Assistant Database" on page 55.

*   "Specifying your Geneticist Assistant Settings" on page 59.

*   "Managing Quality Metrics" on page 77.

Managing Objects in the Geneticist Assistant Database

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. Twelve objects are available in Geneticist Assistant: Chemistry, Instrument, Pathogenicity, Pathogenicity Status, Sample Status, Panel Group, Coverage Region Status, Sample Status, Run Status, Artifact Type, Phenotype, and Ethnicity. Geneticist Assistant is installed with default values for five of these objects: Pathogenicity, Pathogenicity Status, Sample Status, Coverage Region Status, and Run Status. 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	Default Values	Description
Chemistry	No	Refers to the chemistry that was used to prepare the samples for sequencing.
Instrument	No	Refers to the instrument that was used to sequence the samples.
Pathogenicity	Yes	The pathogenicity for a selected variant. Default values are Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign.
Pathogenicity Status	Yes	Indicates the status for changing the pathogenicity for a selected variant. Default values are Pending and Confirmed.
Sample Status	Yes	The status for a sample. Default values are New, QC Passed, Reviewed, and Complete
Panel Group	No	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	Yes	The Coverage Regions 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 tab" on page 200.
Sample Group	No	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.

Object	Default Values	Description
Run Status	Yes	The status for a run. Default values are New, QC Passed, Reviewed, and Complete.
Artifact	No	The artifact type for the sample variant, for example, a Sequencing artifact. A single default value, None, is supplied for artifact type.
Phenotype	No	The phenotype for a patient.
Ethnicity	No	The ethnicity for a run patient, for example, Caucasian (Non-Hispanic).

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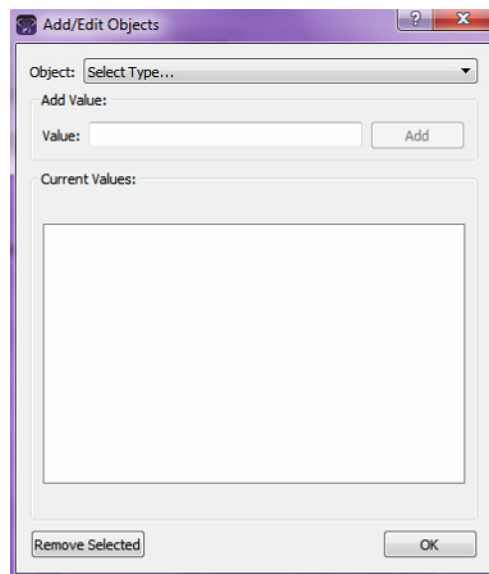
2. On the Object dropdown list, select the object for which you are adding a value.

   If the selected object has default values, or custom values have already been added for the selected object, 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. Do one of the following:

- If you did not add a new pathogenicity value, then go to Step 5.

- If you added a new pathogenicity value, then to correctly order the pathogenicity values from least severe to most severe, select a value in the Current Values pane, and then click Move Up or Move Down as needed. Go to Step 5.

> *If you add a value for pathogenicity, then 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 65.*

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.

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 56.

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

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 56.

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, right-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

When specifying the settings for your Geneticist Assistant installation, if you did not configure the server reference directory before the first time that you logged into Geneticist Assistant, at a minimum, you *must* create your server and client reference directories and specify their locations, and then download reference files to the client reference directory. You *must* also specify the global Quality Control (coverage) settings for your data. See:

- "To configure Geneticist Assistant directories" below.

- "To specify the global Quality Control settings" on page 64.

Optionally, you can also:

- Configure your proxy server settings. See "To configure proxy server settings" on page 61.

- Edit the SoftGenetics license server settings. See "To edit the SoftGenetics license server settings" on page 62.

- Specify an encryption key for patient data. See "To specify an encryption key for patient data" on page 63.

- 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 65.

- Specify the font for column headers and/or column content. See

- Specify the same column display preferences for variants in all the variant panes and variant sub-tabs and/or clear the preferred settings for all the data tabs. See "To apply shared preferences or to clear all tab preferences" on page 69.

- 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 70.

- Specify the requirements for all user passwords for Geneticist Assistant. See "To specify the Geneticist Assistant password requirements" on page 73.

- Specify your third-party settings. See "To specify third-party (Alamut, IGV, OMIM) settings" on page 74.

To configure Geneticist Assistant directories

Before you run Geneticist Assistant for the first time, if you have yet to create your client *reference directory*, which is the directory contains the information for the reference that are used for aligning the data and/or detecting variants, then you must do so. References are stored in their own folders in this directory. If you have yet to configure the server reference directory, which is the directory on the database server that is to host the reference files, then you must also specify this directory location. Optionally, if you have already configured the
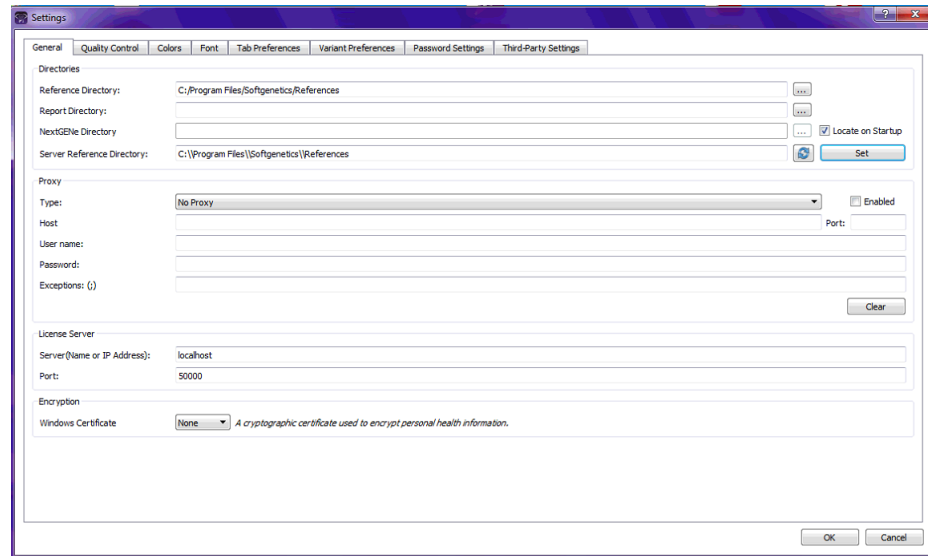
client reference directory, but you have not yet submitted data to the Geneticist Assistant database, then you can change the location of the reference directory. Optionally, you can also change the location of the server reference directory and/or specify a report directory.

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

   The Settings dialog box opens. The General tab is the open tab.

Figure 2-2:   Settings dialog box, General tab

2. Optionally, in the Directories (top) pane, do any or all of the following as appropriate:

   • To specify the location of the client reference directory, or change the location of the directory, next to the Reference Directory field, click the Browse button ⌞...⌟ , and then browse to and select the appropriate directory.

   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.

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

   • To specify the directory in which *all* Geneticist Assistant reports are to be saved, next to the Report Directory field, click the Browse button ⌞...⌟ , and then browse to and select the directory.

   • If your organization also has NextGENe installed, then to use the interactive functionality that is available for Geneticist Assistant and NextGENe, Geneticist Assistant must be able to locate your NextGENe installation. If your Geneticist

Assistant instance is installed on the same client as your NextGENe instance, then leave Locate on Startup selected; otherwise, clear Locate on Startup, and then next to the NextGENe Directory field, click the Browse button ⌊ ... ⌋, and then browse to and select the appropriate directory.

- To specify the location of the server reference directory, or change the location of the current directory, in the Server Reference Directory field, enter the full path to the appropriate directory, and then click Set.

No message opens after you click Set. Also, if you inadvertently clear the path to the server reference directory, then you can click the Refresh icon to immediately repopulate the Server Reference Directory field with the full path and directory name.

3. If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

To configure proxy server settings

If your organization uses a proxy server for external connections, then to view data from external databases such as OMIM or LOVD in Geneticist Assistant, you must configure the settings for the proxy server.

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

The Settings dialog box opens. The General tab is the open tab. By default, the Proxy Type is set to No Proxy.

Figure 2-3:   Settings dialog box, General tab
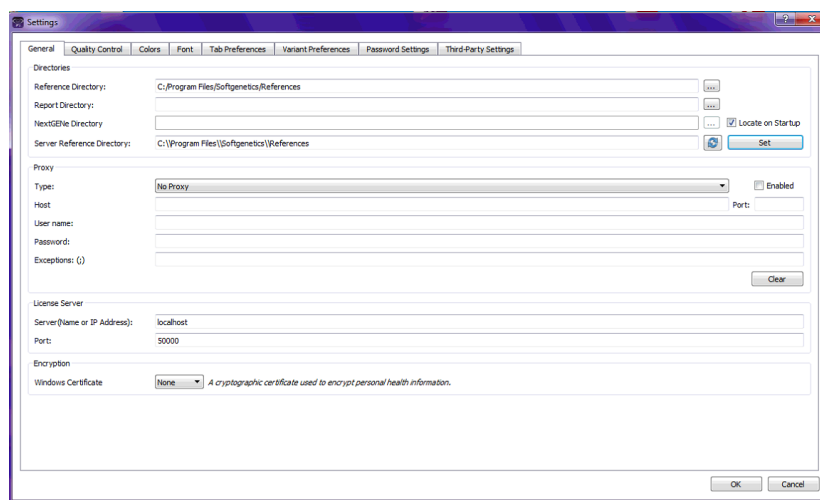
2. On the Type dropdown list, select the proxy type.

3.   In the Proxy pane, configure your proxy server settings.

If you selected DefaultProxy for Type, then the proxy settings for Internet Explorer are used for the connection and you can leave all other proxy settings blank.

4.   If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

To edit 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 24.) If you change the location of this license server, then you must edit the license server settings so that Geneticist Assistant can still connect to the server.

1.   On the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.

The Settings dialog box opens. The General tab is the open tab. The current information (name or IP address) for the SoftGenetics license server is displayed in the Server field.

Figure 2-4:   Settings dialog box, General tab
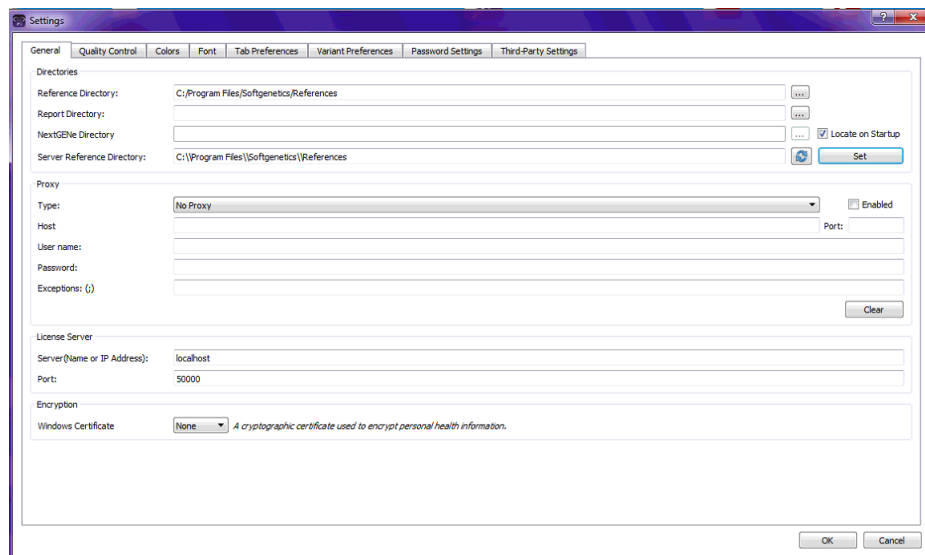
2.   In the License Server pane, in the Server field, enter the fully qualified domain name (FQDN) for the server, or its IP address.

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

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

4. If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

To specify an encryption key for patient data

You can specify an encryption key for the patient data in your Geneticist Assistant database. If encryption is enabled for patient data, then no data is displayed in specific Patient fields such as First Name, Last Name, and so on. Instead, *encrypted* is displayed. To set up encryption for patient data, two files are required:

- Cert.pem file, which you must save in C:\Program Files\SoftGenetics\SoftGenetics Server\ga on the Geneticist Assistant server.

- Cert.pfx, which you can save anywhere on the Geneticist Assistant client. You must double-click this file to launch the Certificate Import Wizard and import the encryption key for your Geneticist Assistant installation.

If your organization already uses encryption for patient data, then these files should already be configured and available for use; otherwise, work with your organization's System Administrator to create and configure these files, or you can contact tech_support@softgenetics.com for assistance.

1. On the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.

   The Settings dialog box opens. The General tab is the open tab.

Figure 2-5:   Settings dialog box, General tab
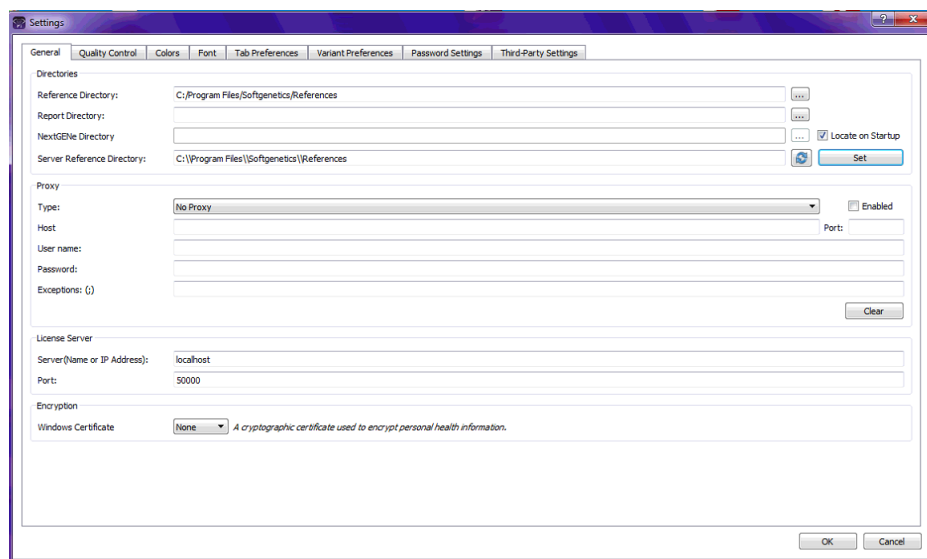
2. In the Encryption pane, select the appropriate Windows certificate.

3. If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

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

    The Settings dialog box opens. The General tab is the open tab.

2. Open the Quality Control tab.

Figure 2-6:   Settings dialog box, Quality Control tab
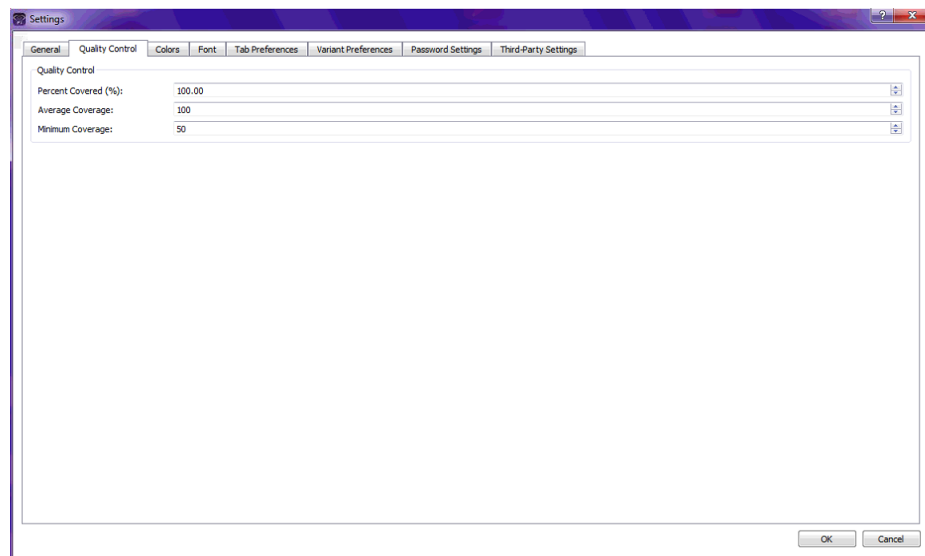
3. Specify the value for the Quality Control settings.

    •   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 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 edit the information on any of the other tabs as needed.

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 various variant sub-tabs and panes in your Geneticist Assistant instance. You can use these default colors, or you can specify other colors on the Colors tab that better meet your business needs.

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

   The Settings dialog box opens. The General tab is the open tab.

2. Open the Colors tab.

   The *first* time that the tab opens, the default highlighting colors for the five system-supplied pathogenicity values are displayed on the tab. If applicable, any custom pathogenicity values are also displayed, but without any highlighting colors. Going forward, the pathogenicity values are highlighted in their specified colors.

   For information about adding a custom pathogenicity value, see "Managing Objects in the Geneticist Assistant Database" on page 55),

Figure 2-7:   Settings dialog box, Default colors shown on the Colors tab
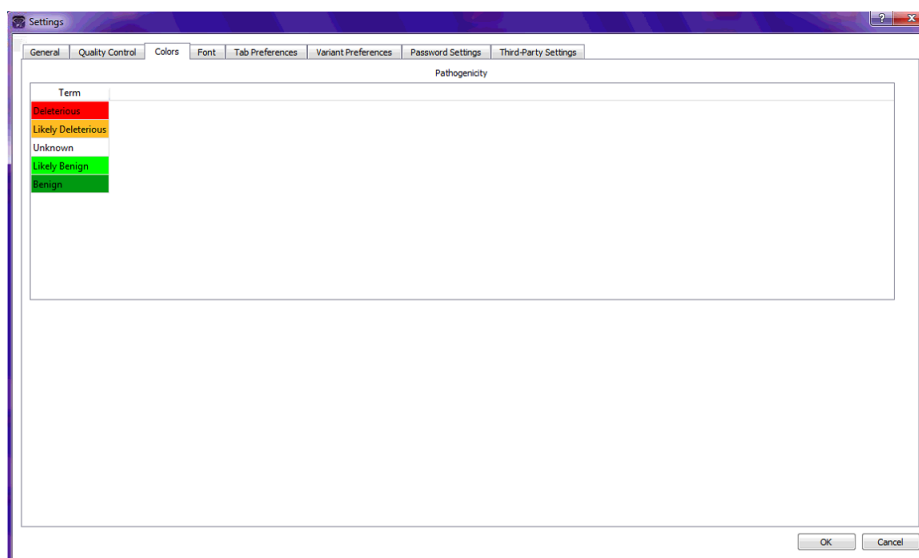
3. Do one of the following:

   • To specify or change the highlighting color for a pathogenicity, double-click the pathogenicity.

   • To specify or change the highlighting color for a case priority, double-click the case priority.

   The Select Highlight Color dialog box opens. The pathogenicity or case priority that you selected is displayed in the title bar of the dialog box. See Figure 2-8 on page 66.

Figure 2-8:   Select Highlight Color dialog box
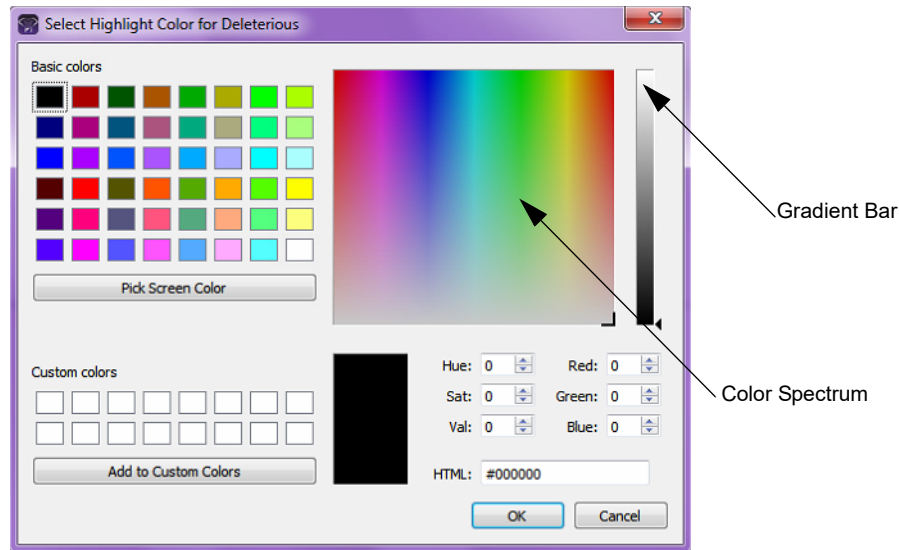
4. Continue to one of the following:

   • "To specify a basic highlighting color for a pathogenicity" below.

   • "To specify a custom highlighting color for a pathogenicity" on page 67.

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 Colors tab on the Settings dialog box remains open. The selected pathogenicity is highlighted in the basic color.

3. Continue selecting pathogenicities or case priorities and specifying basic colors as needed.

4. If you are done 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 edit the information on any of the other tabs as needed.

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 Colors tab on the Settings dialog box remains open. The selected pathogenicity or case priority is highlighted in the custom color.

6. Continue selecting pathogenicities or case priorities and specifying custom colors as needed.

7. If you are done 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 edit the information on any of the other tabs as needed.

To set the font for data column headers and/or content

When Geneticist Assistant is installed, by default, the font for data column headers and data column content is set MS Shelf Dlg2, Normal, Size 8. You can specify a different font, including the style and size, for all your data column headers, for your data column content, or both. You can also apply effects to the font and/or specify the Writing System, which is the operating language for the client on which Geneticist Assistant is installed.

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

   The Settings dialog box opens. The General tab is the open tab.

2. Open the Font tab.

   When the tab opens, Header is selected and the current font information (font, style, and size) for data column headers is displayed. A preview of the selections is displayed in the Sample (lower right) pane. The Writing System is set to the default value of Any.

Figure 2-9:   Settings dialog box, Font tab
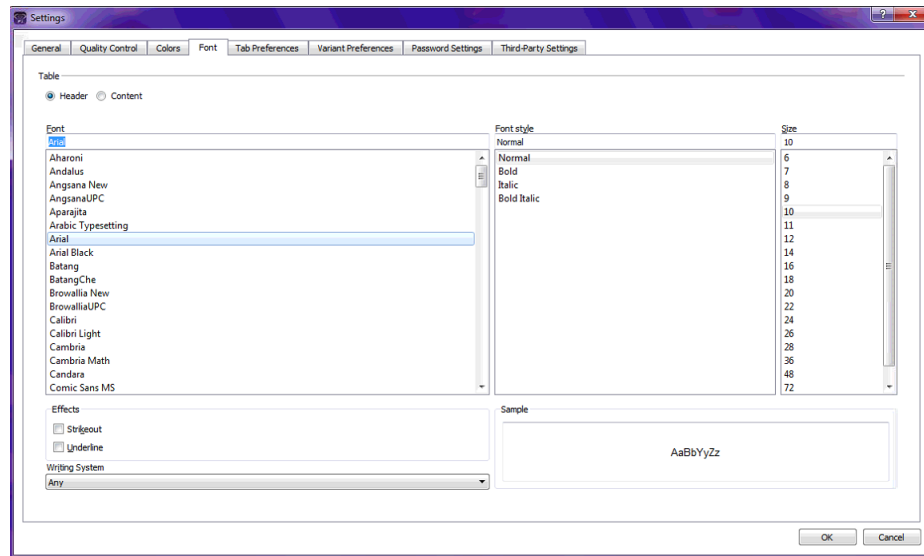
3. Do any or all of the following:

   • To set the font for the data column headers, leave Headers selected, and then choose a different font, font style, and /or size. You can also select or clear a font Effect (Strikeout or Underline).

   • To set the font for the data column contents, select Content, and then choose a different font, font style, and /or size. You can also select or clear a font Effect (Strikeout or Underline).

   • On the Writing System dropdown list, select a different writing system.

4. If you are done 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 edit the information on any of the other tabs as needed.

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 resizing data columns, rearranging the order of data columns, and so on. (See "Setting Display Preferences in the Geneticist Assistant Main Window" on page 31.) 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. *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*, 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 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, or you can clear only the column preferences. Finally, if you have applied a variant filter to a selected sample, then you also have the option of making this filter a persistent filter, which means that if you close and then open the tab again, the filter is "remembered" and is applied again to the sample variants. When you apply a variant filter to a sample, the filter is specific only for this particular sample. Variants of any other sample are not filtered. For example, in Figure 2-10 below, the Variant filter is persistent specifically to Sample 800463.igv.

Figure 2-10:   Variant filter, persistent specifically for a sample

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

    The Settings dialog box opens. The General tab is the open tab.

2. Open the Tab Preferences tab.

    By default, Remember Applied Variant Filters is selected.

Figure 2-11:   Settings dialog box, Tab Preferences tab
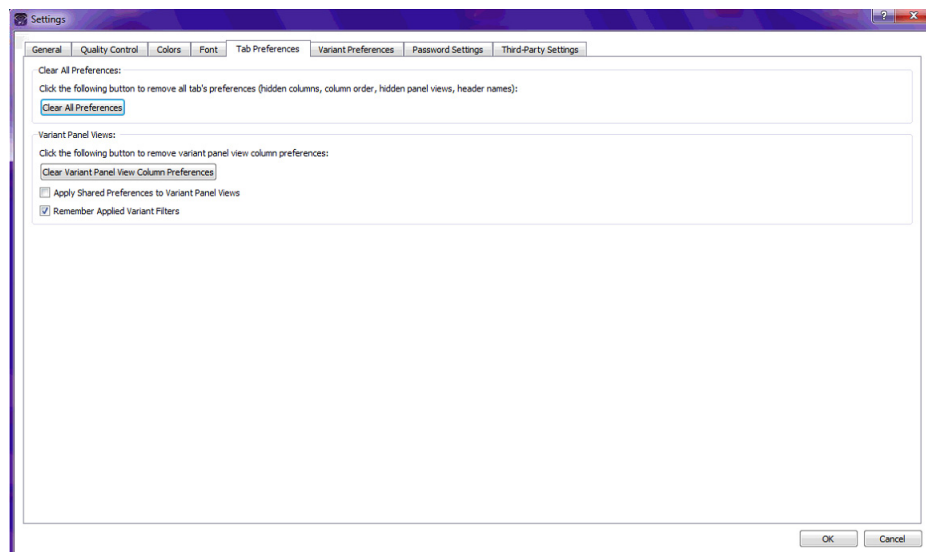
3. Do one of the following:

- To return all tab displays to their default displays (all custom display preferences such as column header names are reset) in a single step, click Clear All Preferences.

- To remove only those column preferences that have been specified for all variant sub-tabs or panes in a single step, click Clear Variant Panel View Column Preferences.

- After you set the column display preferences for a specific variant sub-tab or pane, do one or both of the following:

Option	Description
Apply Shared Preferences to Variant Panel Views.	Select to apply the same preferences that were set for this single variant sub-tab or pane to all the variant sub-tabs or panes on all the other tabs; otherwise, if you clear this option, then you can set display preferences individually for each variant sub-tab or pane.
Remember Applied Variant Filters	Selected by default. Leave selected to make the variant filters persistent for their samples; otherwise, clear this option.

4. If you are done 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 edit the information on any of the other tabs as needed.

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

   The Settings dialog box opens. The General tab is the open tab.

2. Open the Variant Preferences tab. See

Figure 2-12: Settings dialog box, Variant Preferences tab
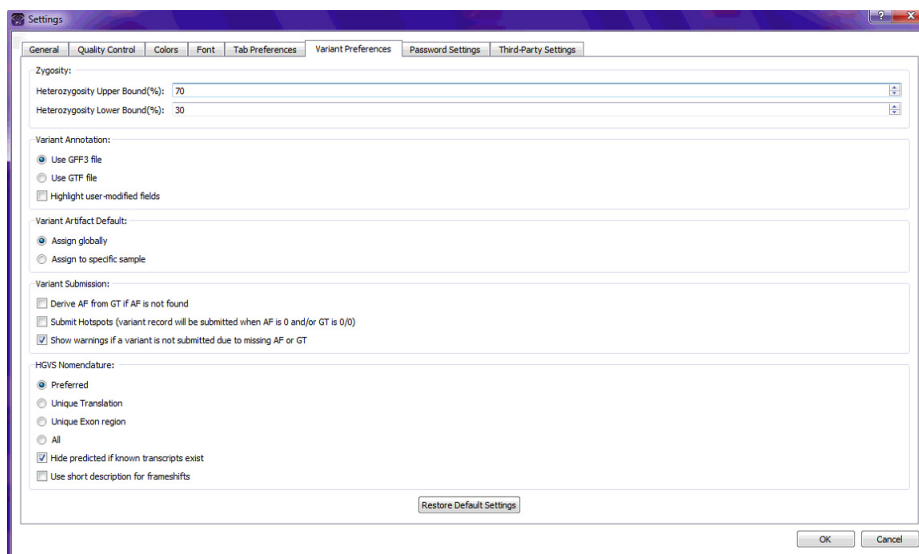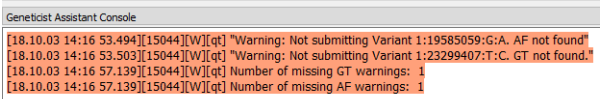
3. Set the variant preferences.

If you edit any variant preferences, then at any time, you can click Restore Default Settings to reset all values to their default values in a single step.

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 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 Geneticist Assistant directories" on page 59.
Highlight user-modified fields	Select this option to highlight any field that a user has modified on all the variant tabs and sub-tabs. This same field is not highlighted on the Variant tab.

Option	Description
Variant Artifact Default	
• Assign globally	• Sets the default option for assigning an artifact type for a variant to assigning the artifact type for all samples. You can edit this default value when assigning the artifact type.
• Assign to specific sample	• Sets the default option for assigning an artifact type for a variant to assigning the artifact type only for the specific sample. You can edit this default value when assigning the artifact type.
Note: The value that you set here determines the default value for Variant Classification on the Update Variant Artifact Type dialog box. See "Patient tab, Variants sub-tab context menu" on page 160, "Variant annotation information" on page 213, and "Sample Variant Data sub-tab context menu" on page 222.	
Variant Submission	
Derive AF from GT if AF not found	Not selected by default. Derive allele frequency (AF) from genotype (GT) if allele frequency not found. If the AF is not found for a variant in the VCF file, then its value is derived from the GT, where: • AF = 0.5 if GT = 0/1, 1/0, 0|1. 1|0 • AF = 1 of GT = 1|1 • AF = 0 if GT = 0|0
Submit Hotspots	Not selected by default. Any variants that are included in a VCF file that have an allele frequency value = 0 and/or genotype = 0/0 in the file *are* submitted to the Geneticist Assistant database.
Show warnings if a variant is not submitted due to missing AF or GT	Selected by default. A warning is displayed in the console if a variant cannot be submitted to the Geneticist Assistant database because the allele frequency (AF) or genotype (GT) is missing for the variant in the VCF file. *Figure 2-13: Sample missing AFGT warning*
HGVS Nomenclature - Determines which transcript is used for the HGVS nomenclature columns if a gene has multiple transcripts.	
• Preferred	Selected by default. Show only a single transcript, either the transcript that is selected by default as the preferred transcript or the transcript that is defined as the preferred transcript in a preferred transcript list. See "To set or delete a preferred transcript list for a panel" on page 110.
• Unique Translation	Show multiple transcripts if the translation varies among the transcripts.
• Unique Exon region	Show multiple transcripts if the exon regions vary among the transcripts.

Option	Description
• All	Shows the HGVS nomenclature based on all the transcripts that are included for the gene.
Hide predicted if known transcripts exist	Selected by default. If known transcripts exist for the gene, then do not include any predicted transcripts in the HGVS columns.
Use short description for frameshifts	NP_000170.1:p.Glu546GlyfsTer16 > NP_000170.1:p.Glu546fs (Long) > (short)

4. If you are done 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 edit the information on any of the other tabs as needed.

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. The only requirement is that the password cannot contain any spaces. You can also specify the following *minimum* requirements for your user passwords: Length, Number of upper and lower case characters, Number of numeric characters, and/or Number of special characters.

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

    The Settings dialog box opens. The General tab is the open tab.

2. Open the Password Settings tab.

    A default value is provided for each password setting.

Figure 2-14:   Settings dialog box, Password Settings tab
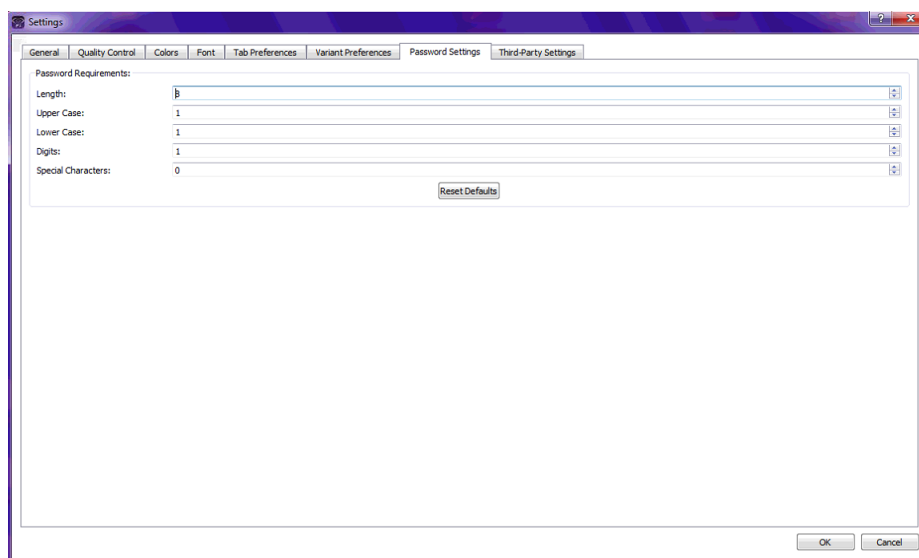
3. Modify any or all of the default values for the user password requirements.

You can click Reset Defaults at any time to reset all password settings to their default values in a single step.

4. If you are done 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 edit the information on any of the other tabs as needed.

To specify third-party (Alamut, IGV, OMIM) settings

You can view variants that were imported in to Geneticist Assistant in Alamut and in the Integrative Genomics Viewer (IGV). You can also view information from OMIM in Geneticist Assistant. You specify the information for these third-party options on the Third-Party Settings tab on the Settings dialog box.

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

   The Settings dialog box opens. The General tab is the open tab.

2. Open the Third-Party Settings tab.

Figure 2-15: Settings dialog box, Third-Party Settings tab
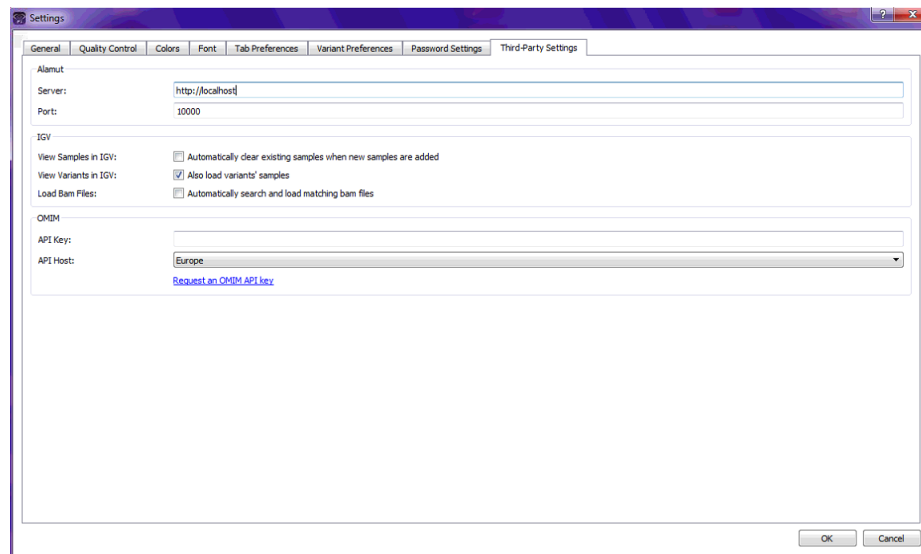
3. Continue to one of the following:

   - "To specify the Alamut settings" on page 75.
   - "To specify the IGV settings" on page 75.
   - "To specify the OMIM settings" on page 76.

To specify the Alamut settings

To view variants that were imported in to Geneticist Assistant in Alamut, the following is required:

- HTTP-based access must be enabled in Alamut. To enable this in Alamut, select Tools > Options > Api, select Enable HTTP-based access, and then set the port to 10000.

- Geneticist Assistant must be able to connect to Alamut and Alamut must be installed on the same computer as the Geneticist Assistant client.

- Alamut must be open to jump to the variant position from Geneticist Assistant.

1. Enter the information for the Alamut server. (The default value is http://localhost).

2. Leave the Port value set to 10000.

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

3. If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

To specify the IGV settings

1. Select or clear your IGV viewing options.

Option	Description
Automatically clear existing samples when new samples are added	When you open a sample in IGV, if other samples are also being displayed, then leave this option cleared to view all samples; otherwise, select it to clear all other samples from the display other than the currently loaded sample.
Also load variants' samples	Selected by default. When viewing a variant in IGV, if you have selected this option, then the sample information for the variant, such as coverage distribution, read pile-up, is also loaded and viewable in IGV.
Automatically search and load missing bam files	If you did not import a BAM file for a sample, then to view the BAM file in IGV, select this option. Geneticist Assistant then looks for the BAM file that matches the VCF file name either in the VCF directory/sub-directory or, if applicable, in the NextGENe project file directory/sub-directory.

2. If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

To specify the OMIM settings

To view information from OMIM in Geneticist Assistant, Geneticist Assistant must be able to connect to OMIM and use an OMIM API key to log you in automatically.

1. In the OMIM (middle) pane, click Request an OMIM API key.

    You are directed to the API Access to OMIM webpage.

2. Complete the necessary information to obtain an OMIM API Access key.

3. Upon receipt of the key, enter the value in the API field.

4. Indicate the connectivity for the API Host (Europe or United States).

5. If you are done 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 edit the information on any this tab or any of the other tabs as appropriate.

Managing Quality Metrics

Quality metrics are crucial in project management, where these metrics are defined as the description of the attributes of the product or project. Every quality metric is an objective number that can be quantified and confirmed. In Geneticist Assistant, you have the option of specifying quality metrics for monitoring and reporting on the NGS data that are you adding to your historical NGS database. In Geneticist Assistant, these quality metrics are grouped in to *metric profiles*. Geneticist Assistant comes pre-populated with three AMP/CAP-recommended metric profiles, with each profile containing the performance metrics that CAP and AMP recommend for monitoring and reporting on NGS bioinformatics pipelines. The majority of these pre-loaded metrics are metrics that are defined in a VCF file.

- AMP/CAP guidelines for all samples

- AMP/CAP guidelines for germline samples

- AMP/CAP guidelines for tumor samples

You can use an AMP/CAP profile as-is, you can add a completely new metric profile (a user-defined profile), you can edit and save an existing user-defined profile as a new profile, or you can import a profile that has been saved in .json format. You can also delete a user-defined metric profile. When you add a new run to the Geneticist Assistant database, you can select the appropriate metric profile for the run, and after the run is complete, the results of this metric monitoring are reported on the Quality Metric Report sub-tab for the Sample tab. (See "Quality Metric Report Sub-Tab" on page 242.)

> *For the relevant article that details these AMP/CAP guidelines, see https://jmd.amjpathol.org/article/S1525-1578(17)30373-2/fulltext.*

Data Source template file

When you are adding a new metric profile, if a pre-loaded metric is one that is commonly defined in a VCF file, then the location of the metric in the sample VCF file is known and the Data Source field on the Manage Quality Metrics dialog box displays this location; however, for the remainder of these pre-loaded metrics, or any user-defined metric, the Data Source field is blank. When you are adding a new metric profile, the locations of all the metrics in the sample VCF file must be explicitly defined. You must create and load a Data Source template file that defines the locations for any pre-loaded metrics that do not have a Data Source value and for all user-defined metrics.

Figure 2-16:   Data Source field indicating metric location
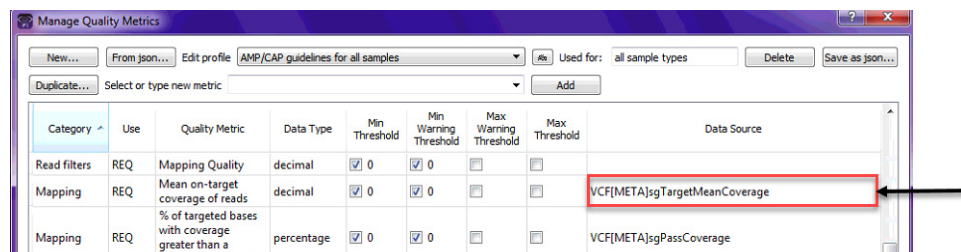
A Data Source template file is simply a sorted VCF file. You load and work with a Data Source template file in the Edit Data Source dialog box.

Figure 2-17:   Edit Data Source dialog box

	Option	Description
1	Quality Metric	Name of the selected quality metric.
2	Category	Category for the selected quality metric (Run, Sample, Variant.)
3	Template File	Name and full directory path to the loaded Data Source template file. To browse to and load a Data Source template file, click the Browse button next to the Template File field.
4	Sample	The name of the sample in the VCF file.
5	Variant	A selected variant in the VCF file.
6	Variant Field/Description	The information that is displayed in this pane depends on the quality metric category (Run, Sample, Variant) that is selected and the VCF file that is loaded. If the data source is not yet defined for a metric, then you can select the appropriate field from this list. **Note:** If the selected metric is not found in the loaded VCF file, then the message "<metric> does not exist in the template file" is displayed.
7	Operators	Operators for creating a formula. Parentheses are also available for setting the order of operations in the formula. If your metric requires combining multiple fields from the VCF file, then you can use these operators to define the metric calculation.
8	() Data Source	Defines how the quality metric is to be derived from a sample VCF file - Primary Data Source, Secondary Data Source, and so on. Click the (+) tab for each data source that is to be used in a calculation.
9	Value	Displays an example value for the metric based on the selected variant.

To add a new metric profile

1. On the Geneticist Assistant main menu, click File > Manage Quality Metrics.

   The Manage Quality Metrics dialog box opens.

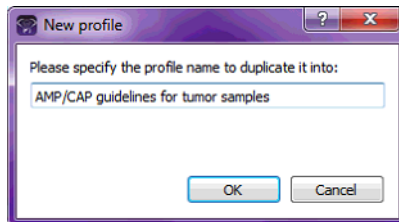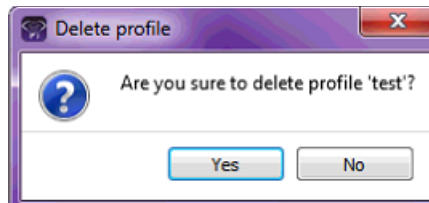*Figure 2-18:   Manage Quality Metrics dialog box*
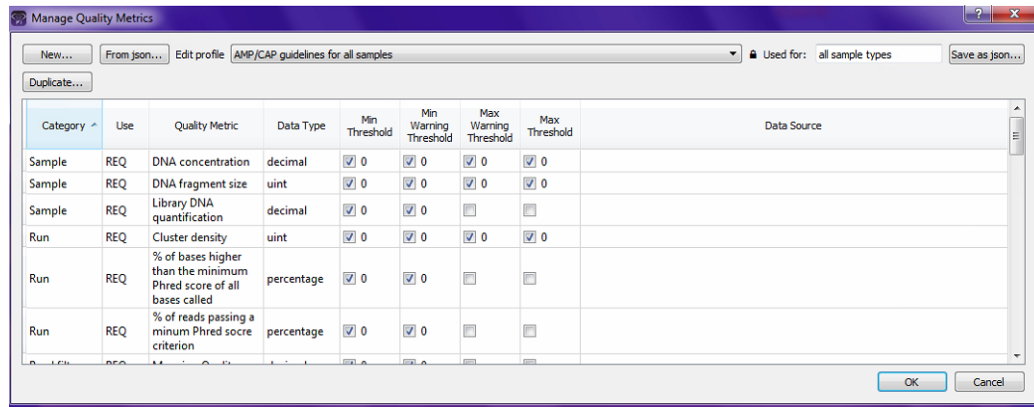
2. Click New.

   The New Metric Profile dialog box opens.

Figure 2-19:   New Metric Profile dialog box
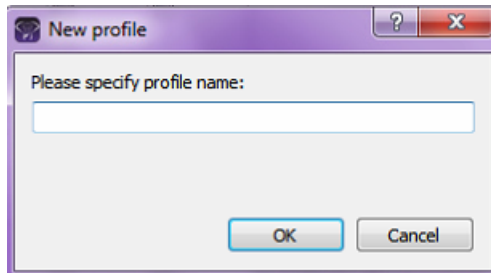
3. Enter a name for the new metric profile, and then click OK.

   The New Metric Profile dialog box closes and the Manage Quality Metrics dialog box remains open. The name of the new profile is displayed in the Edit profile field, and the lower (Metrics) pane is blank.

4. The Used for field identifies the samples types for which the metric profile should be used. When you are creating a new metric profile, the default value is the value for the last loaded metric profile. You can leave this value as-is, or optionally, in the Used for field, enter the sample type for which the profile applies, and then click the Confirm icon that is displayed to the right of the Used for field.

Figure 2-20:   Adding a new metric profile, name and Used for information

As you enter a value in the Used for field, a Reset icon is displayed to the right of the Confirm icon. If you decide that you would rather use the value that had previously been displayed in the field instead of your new value, click the Reset icon.

You must repeat Step 5 through Step 9 for each quality metric that you are adding to the metric profile.

5. In the Select or type new metric field, open the dropdown list and select a pre-loaded metric, or manually enter a metric name. If you manually enter the metric name, you can enter the name for a pre-loaded metric, or you can enter a user-defined metric (one that is not available on the pre-loaded metric list), for example, NextGENe Score. Note the following if you enter the name for a pre-loaded metric:

    *   As you enter the metric name, a dropdown list opens. The list is dynamically updated with the names of the pre-loaded metrics that match the search string that you are entering.

    *   The search string is not case-sensitive, and the string can be found anywhere in the metric name. For example, a search string of "alle" returns **Alle**le fraction, % of bases c**alle**d that are variants, and so on.

6. Click Add.

    *   If the metric is a metric that is commonly defined in a VCF file, then the metric with its attributes (Category, Use, and Data Type) and default threshold placeholders is added to the profile and displayed in the lower (Metric) pane of the Manage Quality Metrics dialog box. The Data Source field displays the location in the sample VCF file where the metric is found. Go to Step 8.

    *   If the metric is a user-defined metric, then you must select the Category, Use, and Data Type values for the metric. You select these values from a list of pre-defined values and you cannot add a new value, or edit the available values. The Use is always set to a default value of REQ (required), but you can select OPT (optional), or OHR (Optiona1but Highly Recommended). The Data Source field is blank. Go to Step 7.

If you are having difficulty locating your user-defined metric in the metric list, remember, you can click a column header to sort the display. See "To change the sort order of data" on page 44.

To remove a metric from the profile, right-click the metric, and on the context menu that opens, click Remove Metric.

7. Double-click in the Data Sources field to open the Edit Data Source dialog box, and then browse to and select the appropriate template file and define the data source.

8. Set the metric thresholds. To set a threshold, make sure that the checkbox for the threshold is selected, and then double-click the threshold placeholder to select it and then change the value.

You can always set a Minimum threshold value, a Maximum threshold value, and/ or Warning threshold values for a metric; however, not all thresholds are applicable for all metrics.

9. If applicable, define the calculation for the metric.

10. Optionally, to save the metric profile in .json format, click Save as json, and then specify a name for the quality metric profile and the location in which to save it.

This option is useful for sharing your metric profiles with other users. You can send this file to Geneticist Assistant users, and they can use the From json function to import the file and use the profile in their Geneticist Assistant instances.

11. Click OK.

The Manage Quality Metrics dialog box closes and the profile is saved to your Geneticist Assistant database. The profile is now available for selection when adding a new run.

To add a new metric profile by copying an existing profile

Instead of adding an entirely new metric profile, you can copy an existing profile, edit the profile, and then save the profile with a new name.

1. On the Geneticist Assistant main menu, click File > Manage Quality Metrics.

The Manage Quality Metrics dialog box opens.

Figure 2-21:   Manage Quality Metrics dialog box
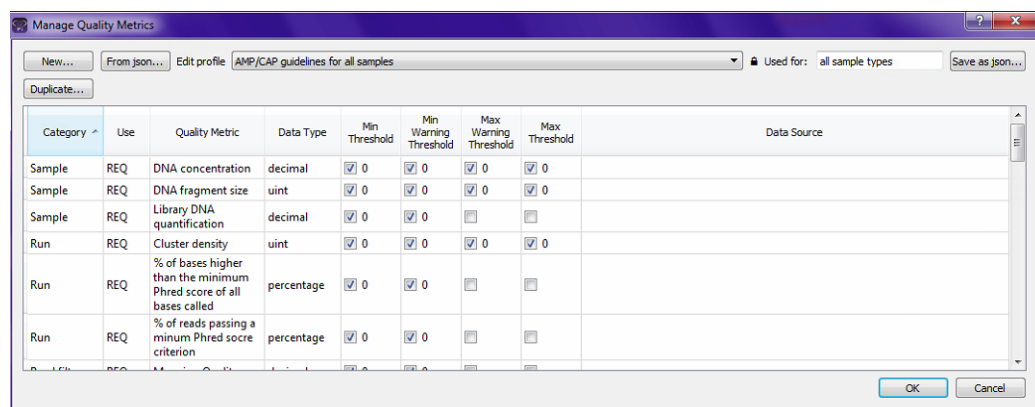
2. On the Edit Profile dropdown list, select the metric profile that you are duplicating and editing.

3. Click Duplicate.

The New Profile dialog box opens.

Figure 2-22:   New Profile dialog box

4. Enter the name for the new metric profile, and then click OK.

The New Profile dialog box closes and the Manage Quality Metrics dialog box remains open. The name of the new profile is displayed in the Edit profile field, and the lower (Metrics) pane is populated with the metrics and thresholds from the copied profile.

5. Edit the profile as needed, and then save the profile. See Step 4 through Step 11 of "To add a new metric profile" on page 79.

To delete a metric profile

You can delete only a non-preloaded metric profile. If you delete a metric profile, any runs that used this profile are not affected. Going forward, the profile is simply not available for selection for a new run.

1. On the Geneticist Assistant main menu, click File > Manage Quality Metrics.

The Manage Quality Metrics dialog box opens.

Figure 2-23:   Manage Quality Metrics dialog box
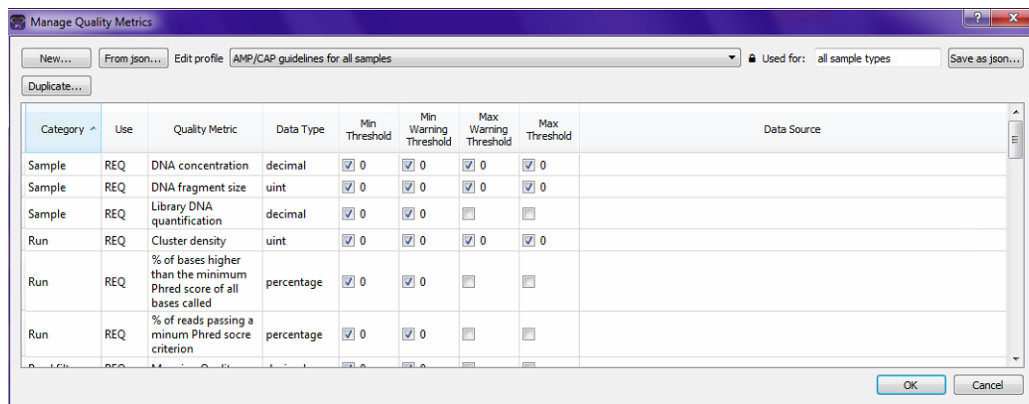
2. On the Edit Profile dropdown list, select the user-defined profile that is to be deleted.

3. Click Delete.

   A Delete profile message opens, asking you if you are sure that you want to delete the profile.

Figure 2-24:   Delete Profile message

4. Click Yes.

   The Delete profile message closes. The Metric Profile is deleted immediately from the Geneticist Assistant database and the Manage Quality Metrics dialog box remains open.

Chapter 3
Geneticist Assistant User Management

In addition to specifying some of the basic settings for Geneticist Assistant, 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 Geneticist Assistant.

This chapter covers the following topics:

- "Managing Users" on page 87.
- "Managing Groups" on page 93.
- "Managing Web Users" on page 98.

Managing Users

Users are the people who log into Geneticist Assistant, whether they are adding and reviewing content, or just using Geneticist Assistant 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 installation, which includes adding new users, deleting users, and editing 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 93.

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 Assistant 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. The User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant installation, their status, and if applicable, their permissions level (Staff or Administrator). See Figure 3-1 on page 88.

Figure 3-1:   User Management dialog box, User tab
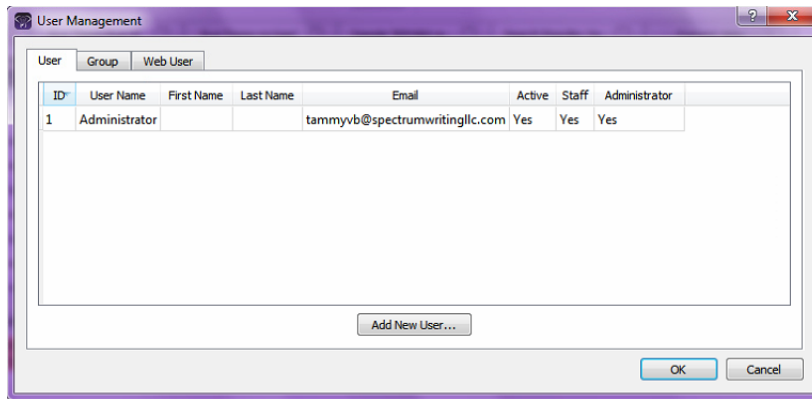
2.   Click Add New User.

The Add New User dialog box opens.

Figure 3-2:   Add New User dialog box
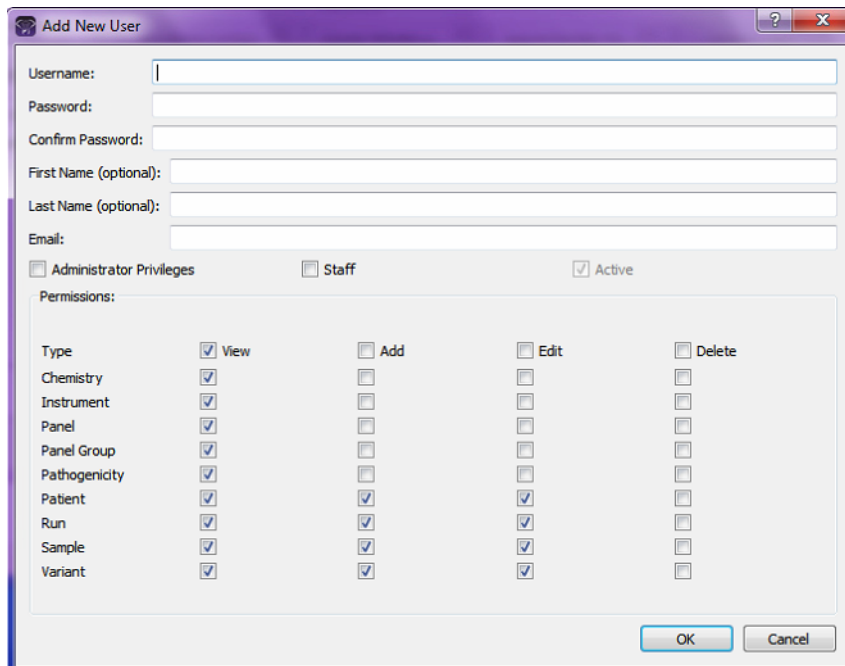
3.   Add the information for the new user.

Field	Description
Username	Required. The login name for the user. Uppercase and lowercase are allowed. No special characters are allowed.

Field	Description
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 73.
Confirm Password	Required. 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 90.

4. In the Permissions pane, add or remove the permissions for the user as needed. You can add or remove 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 remove 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 Add New User dialog box closes. The User Management dialog box remains open.

To delete a user

When you delete a user, the user profile remains in the Geneticist Assistant database. If any comments, changes, and so on are associated with the user, then the username for the deleted user is still shown in the appropriate panes, fields, and so on; however, the user profile is not displayed on any user list in Geneticist Assistant, for example, in the All Users pane on the Add New Group dialog box. To reactivate the user profile, you must add the user again with the same user name.
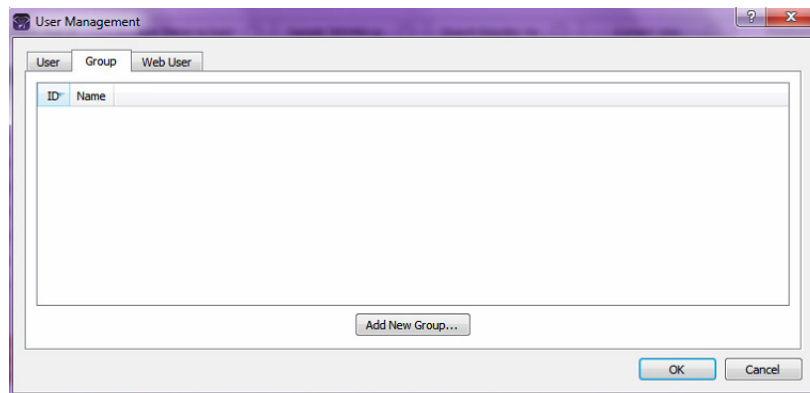
1. On the Geneticist Assistant main menu, click File > User Management.

   The User Management dialog box opens. The User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant installation, their status, and if applicable, their permissions level. See Figure 3-3 on page 90.

Figure 3-3:   User Management dialog box, User tab
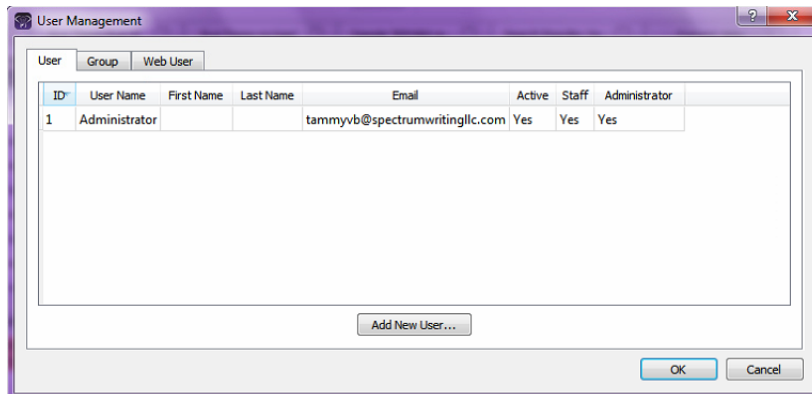
⚠ *No confirmation message opens asking you if you are sure that you want to delete a selected user. Instead, the user is deleted immediately, so be very sure that you want to delete a user before doing so.*

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

    The user is deleted.

To edit a user

When you edit a user, you can edit 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 edit a user name, you must delete the existing user (see "To delete a user" on page 89), 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. The User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant installation, their status, and if applicable, their permissions level.

*Figure 3-4:   User Management dialog box, User tab*
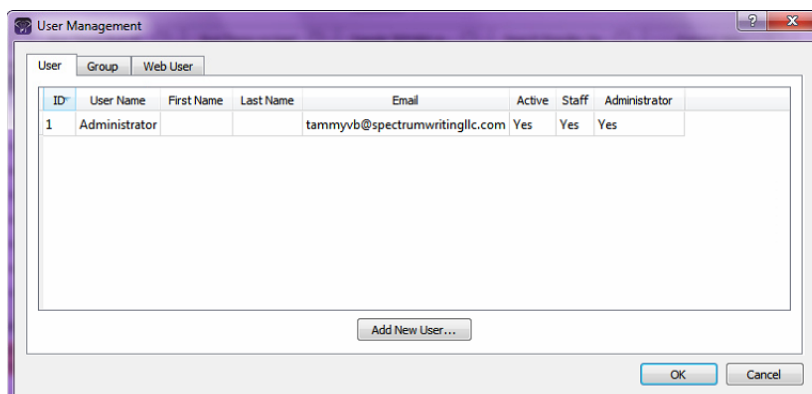
2. Do one of the following:

   - 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. With the exception of the password, the dialog displays all the current information for the user, including the currently assigned permissions.

Figure 3-5:   Edit User dialog box
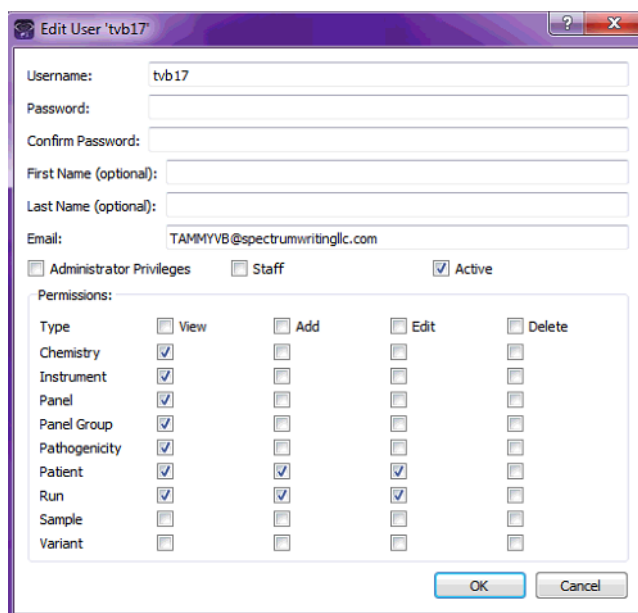
3. Edit any or all of the following information as needed—the user password, the user first and last names, the user email address, the user status (Active), or the user level (Staff or Administrator).

> *If you edit 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. The User tab of the User Management dialog box remain open.

Managing Groups

Instead of assigning permissions to each user on an individual basis, optionally, you can assign users to a *gro*up 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 installation, which includes adding new groups, deleting groups, and editing 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. The User tab is the open tab. See Figure 3-1 on page 88.

2. Click the Group tab to open it.

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

Figure 3-6:   User Management dialog box, Group tab
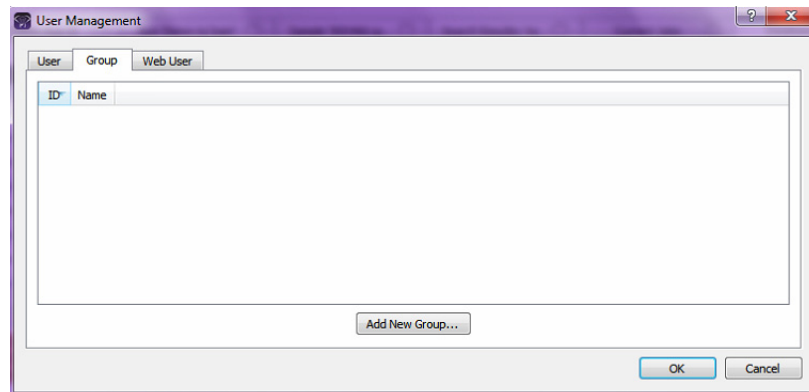
3. Click Add New Group.

   The Add New Group dialog box opens. All users who can be added to a group are displayed in the All Users pane. See Figure 3-7 on page 94.

Figure 3-7:   Add New Group dialog box
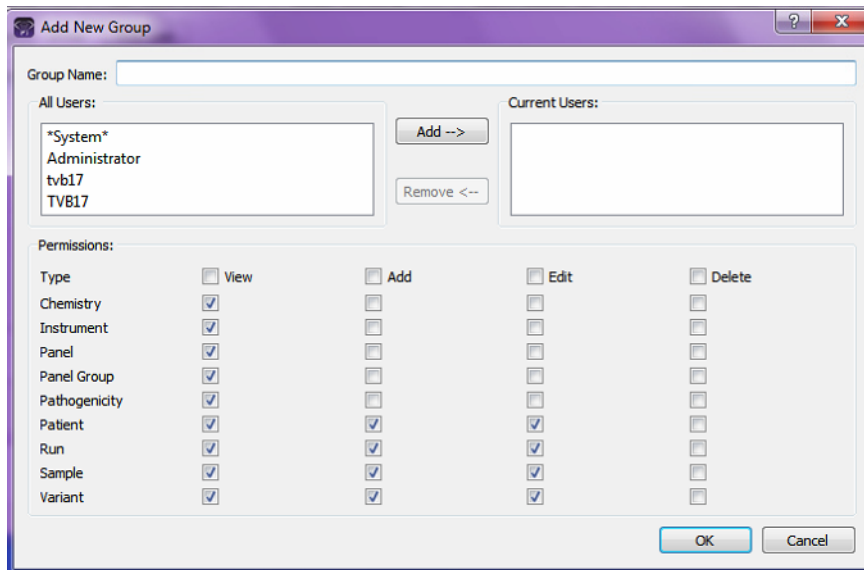
4. Enter the name for the new group.

🖐 *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 (CTRL-click 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 (CTRL-click 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.

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. The User tab is the open tab. See Figure 3-1 on page 88.

2. Click the Group tab to open it.

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

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.

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 95), 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. The User tab is the open tab. See Figure 3-1 on page 88.

2. Click the Group tab to open it.

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

Figure 3-9:   *User Management dialog box, Group tab*
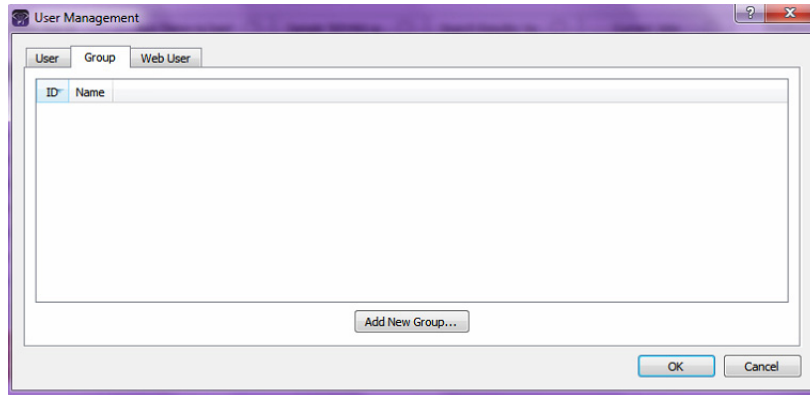
3. Do one of the following:

   • 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.

Figure 3-10:   *Edit Group dialog box*
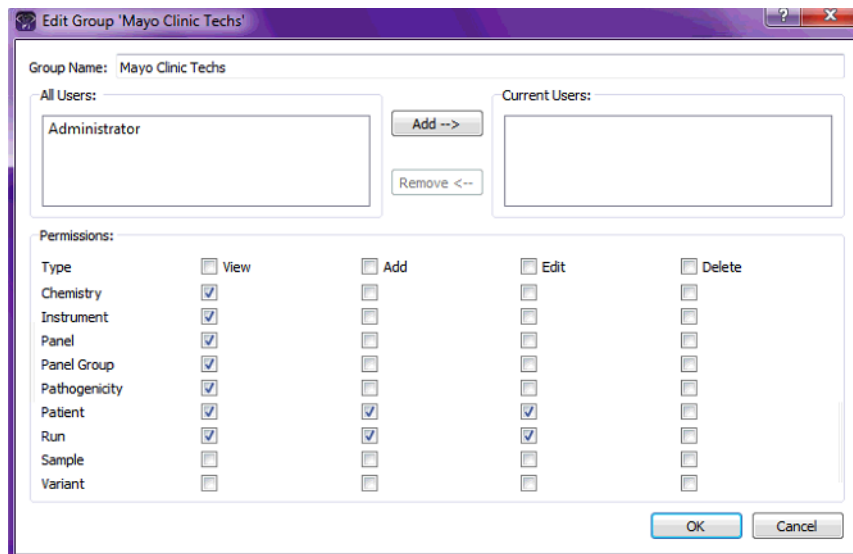
4. To edit the users who belong to the group, do one or both of the following:

   • To add users to the group—In the All Users pane, select the users whom you are adding to the group (CTRL-click 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, (CTRL-click 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.

Managing Web Users

A web user is a user who has login and read/write permissions to the Geneticist Assistant customer portal. A web user can enter your organization's patient and sample information in to the Geneticist Assistant customer portal without ever having to log in to an instance of Geneticist Assistant and accessing your Geneticist Assistant database. As the Geneticist Assistant administrator, you must manage all the necessary web users for the Geneticist Assistant customer portal, which includes adding, deleting, and editing web users.

> *For detailed information about the Geneticist Assistant customer portal, see* *Chapter 12, "The Geneticist Assistant Customer Portal and Submissions," on page 305.*

To add a new web user

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

   The User Management dialog box opens. The User tab is the open tab. See Figure 3-1 on page 88.

2. Click Web User.

   The Web User tab opens. The tab displays all the web users that are currently configured to access your instance of the Geneticist Assistant customer portal.

Figure 3-11: User Management dialog box, Web User tab
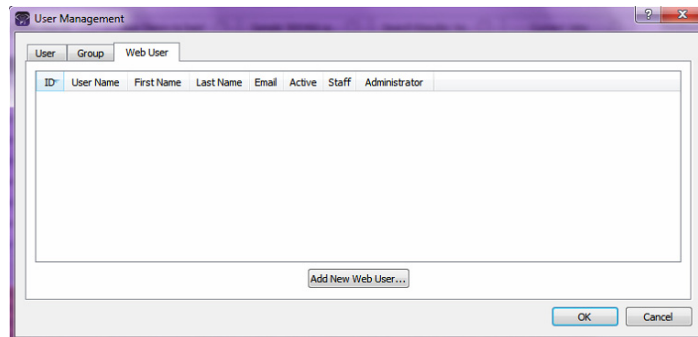
3. Click Add New Web User.

   The Add New User dialog box opens. See Figure 3-12 on page 99.

Figure 3-12: Add New User dialog box (for a web user)
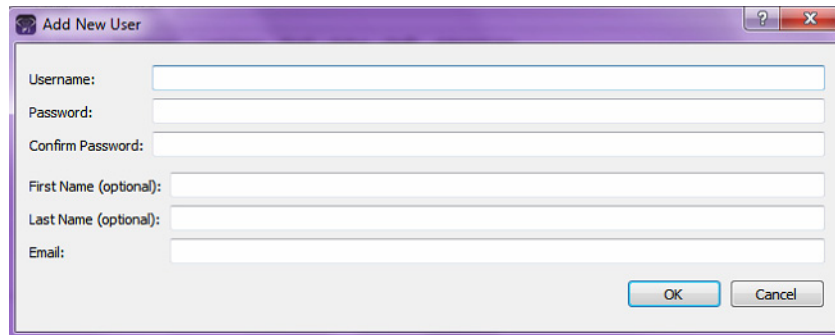
4. Add the information for the new web user.

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 73.
Confirm Password	Required. 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.

5. Click OK.

The Add New User dialog box closes. The User Management dialog box remain open.

When you add a new web user, the user is added with a status of Active, and the user levels of Staff and Administrator are set to No. You cannot edit any of this information.

To delete a web user

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

The User Management dialog box opens. The User tab is the open tab. See Figure 3-1 on page 88.

2. Click Web User.

The Web User tab opens. The tab displays all the web users that are currently configured to access your instance of the Geneticist Assistant customer portal. See Figure 3-13 on page 100.

Figure 3-13:   User Management dialog box, Web User tab
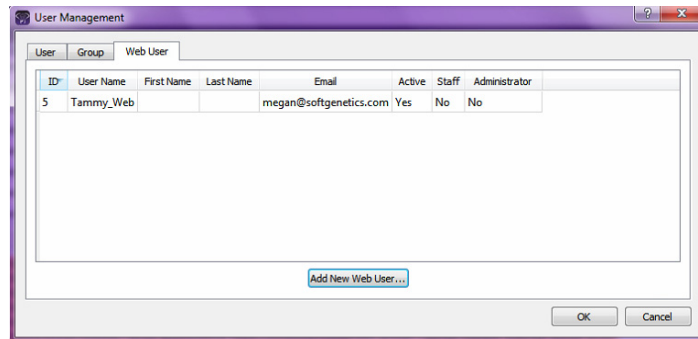
> ⚠️ *No confirmation message opens asking you if you are sure that you want to delete a selected user. Instead, the user is deleted immediately, so be very sure that you want to delete a user before doing so.*

3. Right-click the web user that you are deleting, and on the context menu that opens, click Disable User.

    The web user is deleted.

To edit a web user

When you edit a web user, you can edit the user password, the user first and last names, and the user email address, but you cannot edit the username. To edit a user name, you must delete the existing web user (see "To delete a web user" on page 99), and then add the web user with a new user name.

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

    The User Management dialog box opens. The User tab is the open tab. See Figure 3-1 on page 88.

2. Click Web User.

    The Web User tab opens. The tab displays all the web users that are currently configured to access your instance of the Geneticist Assistant customer portal.

Figure 3-14:   User Management dialog box, Web User tab
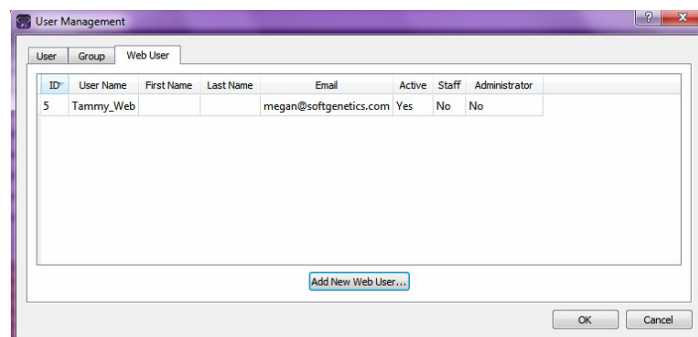
The tab also displays the status (Active) and user levels (Staff and Administrator set to No) for every user. Although this information is displayed for a web user, you cannot edit it.

3. Double-click the web user that you are editing.

   The Edit User dialog box opens. With the exception of the password, the dialog box displays the current information for the selected web user.

Figure 3-15:   Edit User dialog box (for a web user)
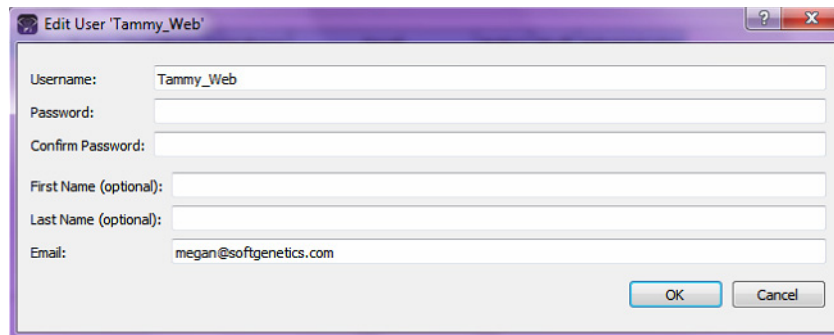
4. Edit any or all of the following information as needed—the user password, the user first and last names, or the user email address.

5. Click OK.

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

Chapter 4
Managing Panels and Subpanels

A *panel* is a collection of genomic regions that are targeted for a specific genomic test or study. You add panels to Geneticist Assistant by providing a BED file that defines the targeted regions, known as *panel regions*. Each sample that you submit to Geneticist Assistant is assigned to a specific panel. Managing panels consists of importing, editing, and deleting panels, and reviewing panel statistics. A *subpanel* contains a fixed number of regions that are contained within a selected panel. To verify coverage for a smaller number of regions that are within the panel that has been selected for a run, you can use a subpanel. Managing subpanels consists of adding, editing, and deleting subpanels.

This chapter covers the following topics:

- "Importing a Panel" on page 105.

- "Editing a Panel" on page 115.

- "Deleting a Panel" on page 117.

- "Reviewing Panel Statistics" on page 118.

- "Adding a Subpanel" on page 119.

- "Deleting a Subpanel" on page 122.

Importing a Panel

A *panel* is a collection of genomic regions that are targeted for a specific genomic test or study. You add a panel to Geneticist Assistant by importing a BED file that defines the targeted regions, known as *panel regions*.

To import a panel

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

   The Manage Panels dialog box opens. The number of panels that have already been imported into your Geneticist Assistant instance is displayed at the bottom of the dialog box. By default, Import from Panel File is selected.

   Figure 4-1:   Manage Panels dialog box
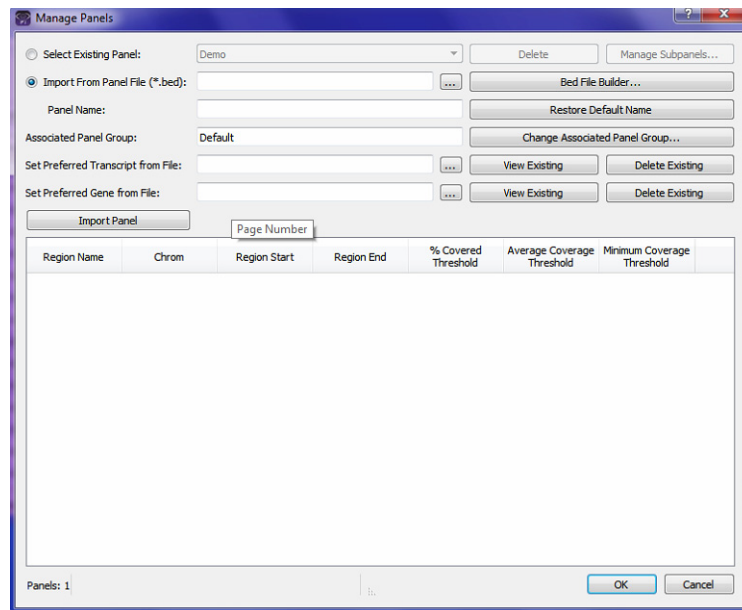
2. Next to Import from Panel File, click the Browse button ⌊...⌋, and then browse to and select the appropriate BED file.

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

   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 for a panel by adding or deleting regions" on page 113), or by clicking Bed File Builder to open the BED File Builder tool and create a BED file that is based on the information that you specify. See "Building a BED File with the BED File Builder Tool" on page 289.

3. 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.

4. If the correct panel group is not selected, then click Change Associated Panel Group to open the Panel Group Association dialog box, and then select a new panel group or click Add New Panel Group to open the Add New Panel Group dialog box and create it.

Figure 4-2:   *Panel Group Association dialog box*
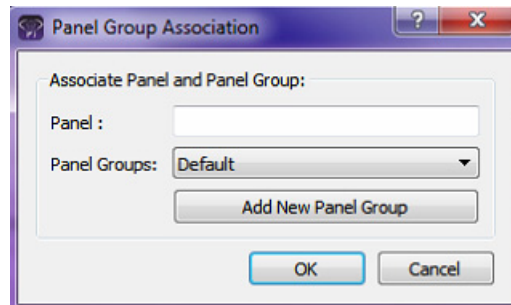
Figure 4-3:   *Add New Panel Group dialog box*
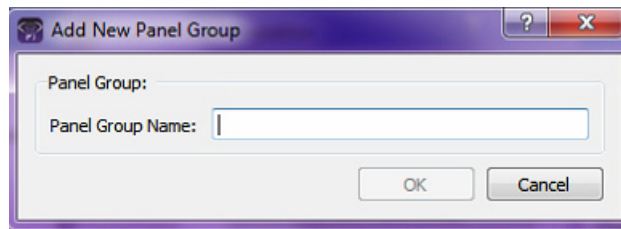
For information about panel groups, see "Managing Objects in the Geneticist Assistant Database" on page 55.

To track variant pathogenicities together for all panels, instead of creating custom panel groups, create a single panel group that is to be used for all panels, for example, a Global panel group.

5. Click Import Panel.

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 64.

6. Do one of the following:

- 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 Current Jobs tab of the Geneticist Assistant main window. The tab shows the progress of importing the new panel. After the panel is successfully imported, the tab is blank.

The time that it can take to import a BED file depends on the size of the BED file. Be patient as some BED files can take over an hour to be imported.

To confirm that the panel has been successfully imported, you can open the Manage Panels dialog box again, choose Select Existing Panel, and make sure that an entry for the imported panel is displayed on the dropdown list of existing panels.
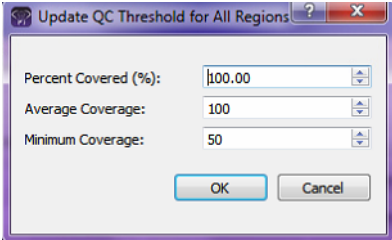
- If you are *not* done with the configuration of the newly added panel, then continue to any of the following:

  - "To manually set new coverage thresholds for panel regions" below.

  - "To create a QC Threshold file for panel regions" on page 108.

  - "To set new coverage thresholds for panel regions with a QC Threshold file" on page 110.

  - "To set or delete a preferred transcript list for a panel" on page 110.

  - "To set or delete a preferred gene for a panel" on page 112.

  - "To create a new BED file for a panel by adding or deleting regions" on page 113.

To manually set new coverage thresholds for panel regions

After you have imported 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 in the Geneticist Assistant database. 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 imported 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 Existing Panel dropdown list, select the appropriate panel.

   c. Click Download and Edit Panel Regions to display the panel regions in the bottom pane of 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 you are editing, 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. Otherwise, continue to any of the following:
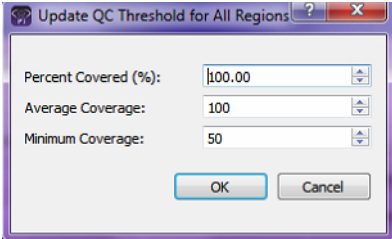
   • "To create a QC Threshold file for panel regions" below.

   • "To set or delete a preferred transcript list for a panel" on page 110.

   • "To set or delete a preferred gene for a panel" on page 112.

   • "To create a new BED file for a panel by adding or deleting regions" on page 113.

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 thresholds to a different panel.

1. If you have just imported 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 4-5: Update QC Threshold for All Regions dialog box* 2. For each coverage value that you are editing, 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 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 BED file for the panel. 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

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 create a new BED file for a panel by adding or deleting regions" on page 113.

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 (which saves these values in the Geneticist Assistant database), and then load this text file to apply these coverage values to a different panel.

1. If you have just imported 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 108.

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. Otherwise, continue to one or both of the following as needed:

   - "To set or delete a preferred transcript list for a panel" on page 110.

   - "To set or delete a preferred gene for a panel" on page 112.

   - "To create a new BED file for a panel by adding or deleting regions" on page 113.

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 Existing 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.

Figure 4-6:   Preferred Transcript dialog box
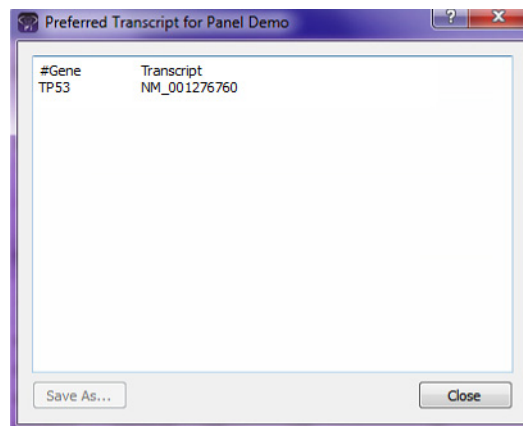
    •   To save the preferred transcript list as a text (.txt) file, click Save As in the Preferred Transcript dialog box; otherwise, click Close to close the dialog box.

        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, then next to the Set Preferred Transcript from File field, click the Browse button [...] 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 click 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 107.

- "To create a QC Threshold file for panel regions" on page 108.

- "To set new coverage thresholds for panel regions with a QC Threshold file" on page 110.

- "To set or delete a preferred gene for a panel" below.

- "To create a new BED file for a panel by adding or deleting regions" on page 113.

To set or delete a preferred gene for a panel

If a variant is found at a position where there are overlapping genes, then you can specify the gene that is to be used for the panel. The file that specifies the preferred gene is a text file that contains the complete gene name on a single line.

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 Existing Panel dropdown list, select the appropriate panel.

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

- To view the genes that are currently set as the preferred genes for the panel, click View Existing.

   The Preferred Gene for <Panel Name> dialog box opens. The dialog box displays the genes that are currently set as the preferred genes for the panel.

Figure 4-7:   Preferred Gene dialog box
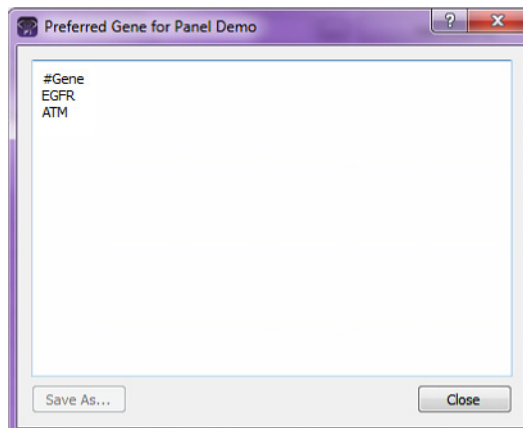
- To save the preferred gene file as a text (.txt) file, click Save As in the Preferred Gene dialog box; otherwise, click Close to close the dialog box.

  The Manage Panels dialog box remains open.

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

- To set preferred genes for the panel, then next to the Set Preferred Gene from File field, click the Browse button [...] and in the Open File dialog box, browse to and select the appropriate preferred gene file, and then click Open.

  The Open File dialog box closes. The full path to the selected preferred gene file is displayed in the Set Preferred Gene from File field.

- To delete the preferred genes for the panel, click Delete Existing, and then click 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 107.
   - "To create a QC Threshold file for panel regions" on page 108.
   - "To set new coverage thresholds for panel regions with a QC Threshold file" on page 110.
   - "To set or delete a preferred transcript list for a panel" on page 110.
   - "To create a new BED file for a panel by adding or deleting regions" below.

To create a new BED file for a panel 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 a BED file for a panel, you can add regions from an existing BED file to it and then save the combined regions as either a new BED file or a panel. You can also delete regions from the loaded BED file and save the modified regions as a new BED file or panel.

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 imported 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 Add Regions from File. 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.
To delete regions from the loaded BED file	1. Click the region that you are deleting. (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 panel does not delete the panel. To delete the panel itself, see "Deleting a Panel" on page 117.

3. On the Manage Panels dialog box, click Save Panel Regions to 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 107.
- "To create a QC Threshold file for panel regions" on page 108.
- "To set new coverage thresholds for panel regions with a QC Threshold file" on page 110.
- "To set or delete a preferred transcript list for a panel" on page 110.
- "To set or delete a preferred gene for a panel" on page 112.

Editing a Panel

Managing panels includes editing panels that will be used for studying your data in Geneticist Assistant.

To edit a panel

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

    The Manage Panels dialog box opens.

Figure 4-8:   Manage Panels dialog box
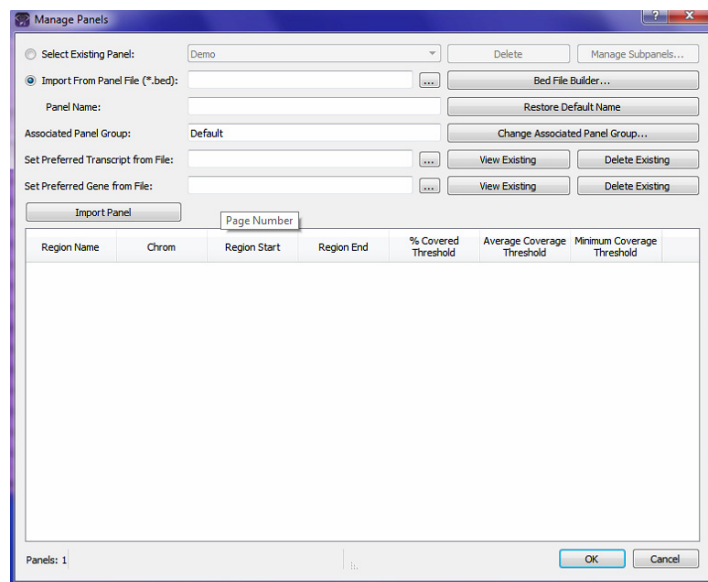
2. On the Select Existing Panel dropdown list, select the panel that you are editing.

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.

Figure 4-9:   Panel Group Association dialog box
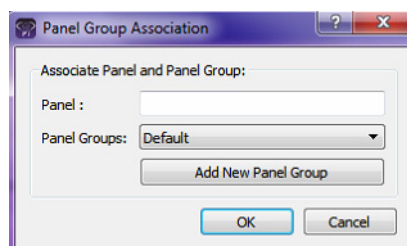
- To manually set new coverage thresholds for the panel regions, see "To manually set new coverage thresholds for panel regions" on page 107.

- 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 110.

- 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 110.

- To set a preferred gene for the panel, see "To set or delete a preferred gene for a panel" on page 112.

- To create a new BED file for the panel, see "To create a new BED file for a panel by adding or deleting regions" on page 113.

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 panel, not the entire panel, see "To create a new BED file for a panel by adding or deleting regions" on page 113.

To delete a panel

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

   The Manage Panels dialog box opens.

Figure 4-10: Manage Panels dialog box
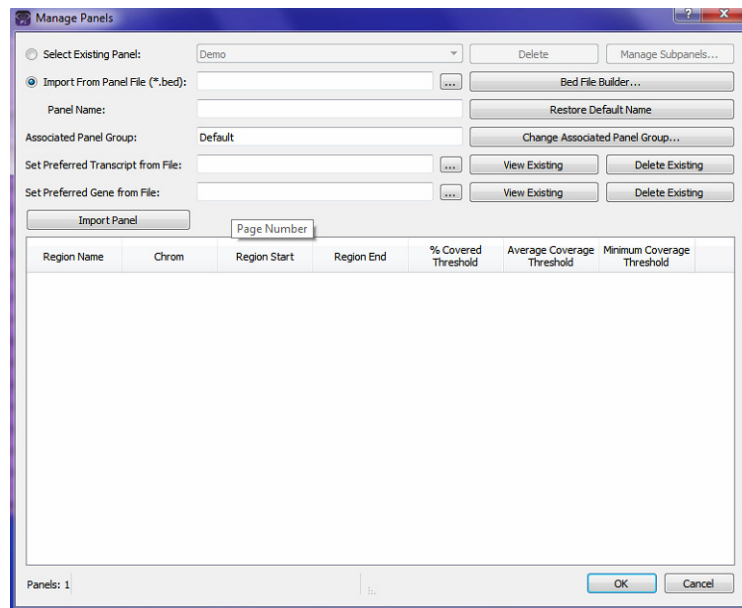
2. On the Select Existing Panel dropdown list, select the panel that you are deleting.

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. By default, all samples for the panel are included. You can edit the options for displaying panel statistics such as setting specific date range, or selecting specific samples or runs.

To review panel statistics

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

   The Panel Statistics tab opens.

2. On the Panels dropdown list, select the panel for which the statistics are to be viewed.

3. Change any of the display options for the Panel Statistics tab, and then click OK to load the panel statistics based on the specified options.

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 Runs tab, the Samples sub-tab on the Patient tab, the Run tab, or the Variant tab. For a complete description of this tab, see "Reviewing Panel Statistics" on page 319.

Adding a Subpanel

A *subpanel* contains a fixed number of regions that are contained within a selected panel. To verify coverage for a smaller number of regions that are within the panel that has been selected for a run, you can use a *subpanel*. Managing panels includes adding the new subpanels that are to be used for targeting a specific genomic test or study, and creating a new BED file for a subpanel.

To add a subpanel

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

    The Manage Panels dialog box opens. See Figure 4-1 on page 105.

2. On the Select Existing Panel dropdown list, select the panel from which the subpanel is to be created.

3. Click Manage Subpanels.

    The Manage Subpanels dialog box opens. The first time that the dialog box opens, all fields are blank; otherwise, it displays the information for the subpanel that was last viewed and/or modified.

Figure 4-11:   Manage Subpanels dialog box
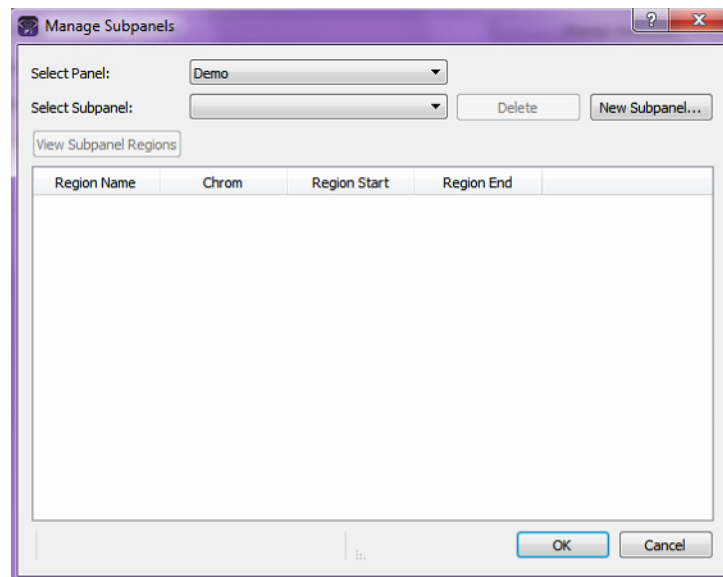
4. Select the panel for which you are adding the subpanel.

5. Click New Subpanel.

The Add Subpanel dialog box opens.

Figure 4-12:   Add Subpanel dialog box
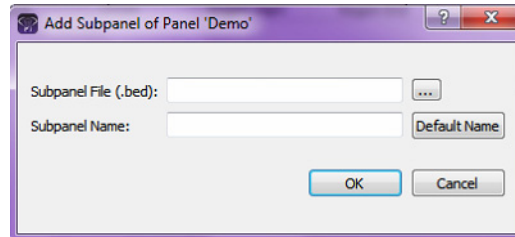
6. Next to the Subpanel File (.bed) field, click the Browse button ⸤...⸥ , and then browse to and select the BED file for the subpanel that you are adding.

The Subpanel file field is populated with the full directory path to the selected BED file, and by default, the subpanel name is set to the name of the selected BED file.

> *If the correct BED file is not available for a subpanel, then you can use 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 289.*

7. Optionally, edit the name for the subpanel.

> *You can click Default Name to reset the BED file name to the default value.*

8. Click OK.

The Add Subpanel dialog box closes. The Manage Subpanels dialog box remains open. The information for all the regions (Region Name, Chromosome Number, Region Start, and Region End) is displayed in the lower pane of the dialog box.

9. If you are done with the configuration of the newly added subpanel, then click OK to close the Manage Subpanels dialog box and return to the Manage Panels dialog box; otherwise, continue to "To create a new BED file for a subpanel by adding or deleting regions" below.

To create a new BED file for a subpanel by adding or deleting regions

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

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 subpanel, then the Manage subpanels dialog box is already open and all the panel regions for the subpanel 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. Choose Select existing panel, and then on the Panel dropdown list, select the appropriate panel.

   c. Click Manage Subpanels.

   d. On the Manage Subpanels dialog box, select the appropriate subpanel.

   e. Click View subpanel regions.

2. Do one or both of the following as needed:

Action	Steps
To add regions from an existing BED file	1. Next to the Add Regions from BED File field, click the Browse button [...]. 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 you are deleting. (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 subpanel. To delete the subpanel itself, see "Deleting a Subpanel" on page 122.

3. On the Manage Subpanels 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 in the following format: <subpanel>_<Panel> and it is saved in the same directory as the loaded BED file. You can change one or both of these values.

4. Click OK.

   The regions are saved as a subpanel.

Deleting a Subpanel

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

*When you delete a subpanel, you delete the entire subpanel. To simply delete
regions from a BED file, not the entire subpanel, see "To create a new BED file for
a subpanel by adding or deleting regions" on page 120.*

To delete a subpanel

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

    The Manage Panels dialog box opens. See Figure 4-1 on page 105.

2. Choose Select existing panel, and then on the Panel dropdown list, select the appropriate
    panel.

3. Click Manage Subpanels.

    The Manage Subpanels dialog box opens.

Figure 4-13: Manage Subpanels dialog box
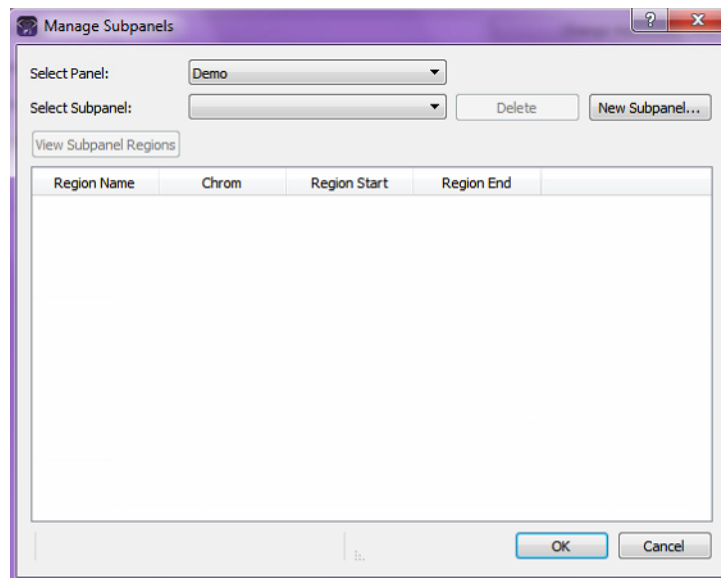
4. Select the subpanel that you are deleting.

5. Click Delete.

A Delete Subpanel message opens, asking you if you are sure that you want to delete the selected subpanel from the database.

Figure 4-14:   Delete Subpanel message
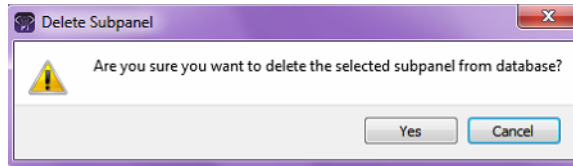
6. Click Yes.

The Delete Subpanel message closes, and a Subpanel Deleted message opens, indicating that the selected subpanel was successfully deleted.

Figure 4-15:   Subpanel Deleted message
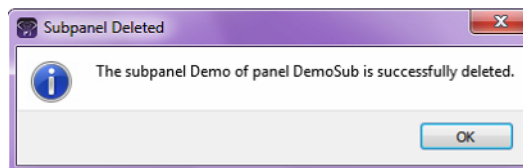
7. Click OK.

The Subpanel Deleted message closes. The Manage Subpanels dialog box remains open. The subpanel and its regions are no longer displayed in the dialog box.

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 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.

Geneticist Assistant supports only sorted VCF files.

This chapter covers the following topics:

- "Adding Patients" on page 127.
- "Accessing and Reviewing Patient Records" on page 137.
- "Adding a Run" on page 170.

For information about managing your run settings (chemistries, instruments, and so on), see "Managing Objects in the Geneticist Assistant Database" on page 55.

Adding Patients

In addition to managing settings (chemistries, instruments, and so on) for your runs, you must 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 or Customer Poral. You have three options available for adding a new patient. You can:

- Manually add one patient at a time.

- Import a Patient batch file to add multiple patients to the database in a single step.

- Import a Submissions batch file to add multiple patients to the Customer Portal 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, or even create the patient record if needed, and then associate the sample with the record. 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 one patient at a time

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

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

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

Figure 5-1:   Add New Patient dialog box, Add Patient tab
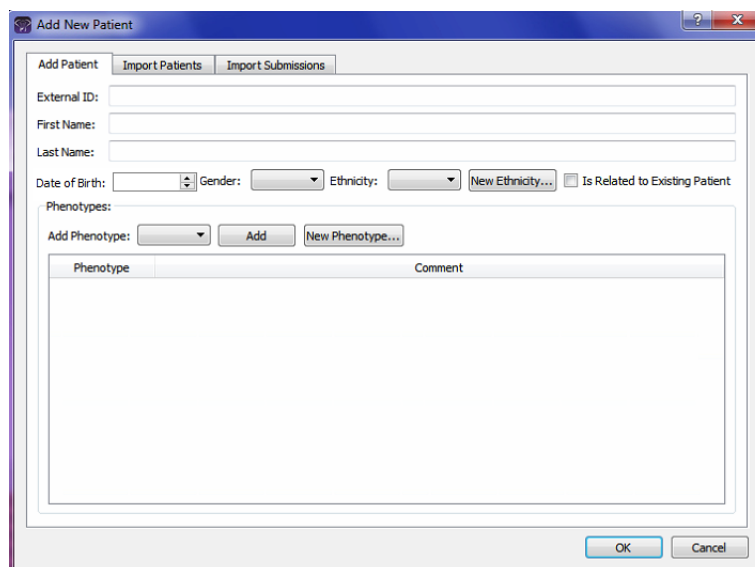
2. Enter the information for the new patient.

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.
Date of Birth	The patient's date of birth. You must click in the field, and then manually enter a new value, or you can use the Up/Down arrows.
Gender	Male or Female.
Ethnicity	The ethnicity for a patient, for example, Caucasian (Non-Hispanic). **Note:** If the appropriate ethnicity is not available, then you can click New Ethnicity and add it. You can also add ethnicities using the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55.

3. If applicable, to specify relationships for the new patient, select Is Related to Existing Patient and then do the following:

   a. Select the relationship type, for example, Father. (The available relationship options depend on the patient's gender.)

   b. 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.)

   c. Click Add.

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

Figure 5-2: Add New Patient dialog box, Relationship options

Geneticist Assistant User's Manual
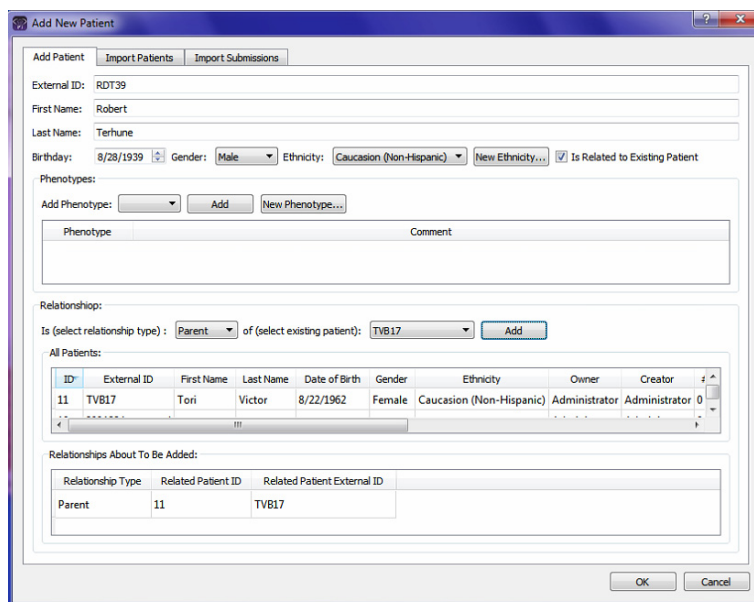
4. Repeat Step 3 to add as many relationships for a patient as needed.

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

5. If applicable, to add a phenotype for the patient, do any of the following:

   • On the Add Phenotype dropdown list, select the phenotype, and then click Add. You can repeat this step as many times as needed to add multiple phenotypes for the patient.

      The added phenotype is displayed in the Phenotype pane.

   • If the appropriate phenotype is not available, then you can click New Phenotype to open the New Phenotype dialog box and enter the phenotype. As you enter the search string, a dropdown list opens. The list is dynamically updated with the phenotypes that are found in the Human Phenotype Ontology database that match the search string.

> *The search string is not case-sensitive, and the string can be found anywhere in the search results. For example, a search string of "thyroid" returns phenotypes of **thyroid** adenoma and secondary hyper**thyroid**oism.*

Figure 5-3:   New Phenotype dialog box
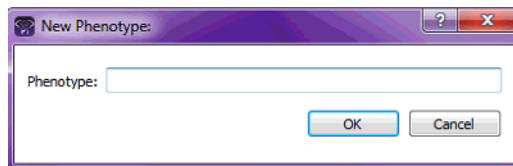
> *You can also add phenotypes using the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55.*

6. Optionally, to add a comment for the phenotype, do one of the following; otherwise, go to Step 7.

   • In the Comment field, single click twice, enter the comment, and then click anywhere outside the field.

   • Right-click the phenotype and on the context menu that opens, click Add Patient Phenotype Comment.

7. Click OK.

   Two results are possible:

   • If you have encryption enabled for your Geneticist Assistant instance, then the Add New Patient dialog box closes. The new patient record is displayed on the Patients tab. For specific identifying information, asterisks (*) are displayed.

• If you do not have encryption enabled for your Geneticist Assistant instance, then an Invalid Encryption Certificate Setting dialog box opens, warning you that no Encryption Certificate is selected and that data will be submitted without encryption. Go to Step 8.

Figure 5-4:   Invalid Encryption Certificate Setting dialog box
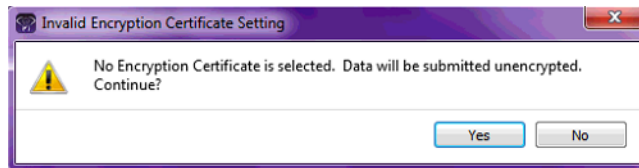
8. Click Yes.

   Both the Invalid Encryption Certificate Setting dialog box and the Add New Patient dialog box close. The new patient record is displayed on the Patients tab, with all identifying information fully visible.

To import a Patients batch file

You can import a Patients batch file to add multiple patients in a single step to the Geneticist Assistant database. The Patients batch file must be either a comma-separated values (.csv) file, a tab-delimited text (.txt) file, or a tab-separated values (.tsv) file. The first line of the file must be the pound/hashtag (#) symbol, followed by delimited column headers. The file *must* contain the following columns of information in the order listed—the External ID, the patient last name, the patient first name, the patient date of birth, and the patient gender. Multiple phenotypes and multiple relationships must be separated by semi-colons. In addition to the required columns, the Patients batch file can include other optional standard fields, such as ethnicity, phenotypes, and relationships, as well as custom fields. Patient relationships must be in the format ExternalID;Relationship, for example, ABS17;Father. When importing the patients, you can opt to add any custom patient fields to the Geneticist Assistant database. Figure 5-5 below is an example file that shows the required column order and column names for a Patients batch file. The example file also includes custom data columns.

To view and edit the custom fields for a patient, you must edit the patient record (Edit Patient). See "To use the context menu for a patient (Patients tab)" on page 141.

Figure 5-5:   Patients batch file with required columns and custom unmatched columns

ernal ID	Last Name	First Name	Date of Birth	Gender	Ethnicity	Phenotype	Relationships	Sample type	Hair Color	Eye
1320476	Doe	John	5/6/1913	Male	Causcasian			Tissue	Brown	Brow
1320683	Doe	Jane	3/2/1920	Female	Causcasian			Tissue	Blonde	Blue
1315487	Doe	Jill	6/10/1946	Female	Causcasia	phe1	1320476;Child;1320683;Child	Tissue	Brown	Brow
800402	Doe	Joan	6/10/1946	Female	Causcasia	phe1	1320476;Child;1320683;Child	Blood	Brown	Blue

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

   The Add New Patient dialog box opens. The Add Patient tab is the opened tab. See Figure 5-1 on page 127.

2. Click the Import Patients tab to open it.

Figure 5-6:   *Add New Patient dialog box, Import Patients tab*
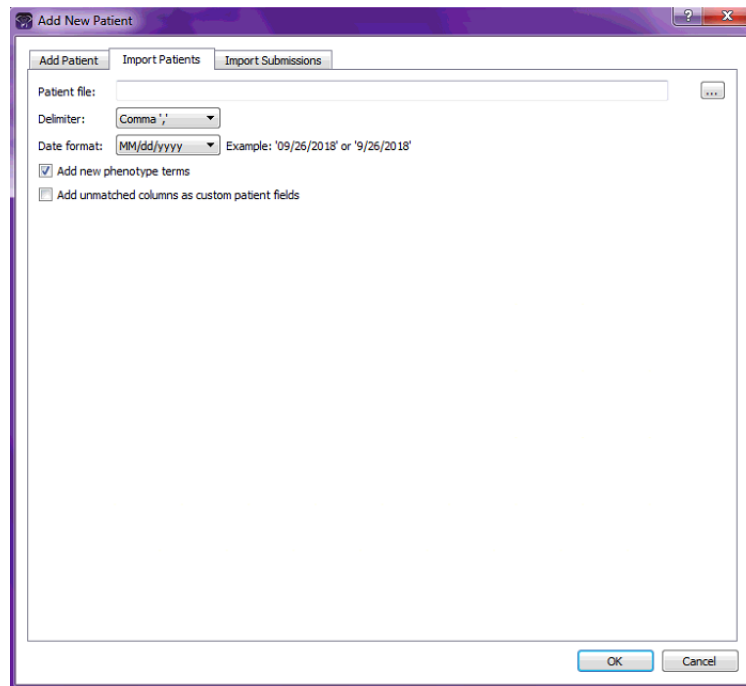
3. Next to the Patient File field, click the Browse button ⬚, and then browse to and select your patient file.

4. Specify the properties of your patient file.

Property	Description
Data Delimiter	Indicate the delimiter used (Comma or Tab).
Date of Birth Format	Select the format used for the patients' dates of birth.
Add New Phenotype Terms	Selected by default. If the patient file contains phenotypes in the Phenotypes column that have not already been added to the Geneticist Assistant database through either the Manage Objects function (see "Managing Objects in the Geneticist Assistant Database" on page 55), or the Add Phenotype option on the Add Patient tab (see "Patient tab, Patient sub-tab, Phenotype pane" on page 148), then leave this option selected to automatically add these phenotypes to the database. Otherwise, clear this option.

Property	Description
Add unmatched columns as custom patient fields	In addition to the required information for the Patients batch file, the file can include any other information as needed, for example, Sample Type, Weight, Age at Death, and so on. Select this option to include this information in the Geneticist Assistant database for the patients.

5. Click OK.

You results are determined by whether you have enabled encryption for your patient data. Continue to one of the following:

*   "To add patients to the Geneticist Assistant database with encryption enabled" below.

*   "To add patients to the Geneticist Assistant database with encryption enabled" on page 132.

The results below illustrate the most common scenarios for adding a new patient to the Geneticist Assistant database, and are meant only as guidelines for adding patients, and if applicable, custom fields, to the Geneticist Assistant database.

To add patients to the Geneticist Assistant database with encryption enabled

If you have encryption enabled for your Geneticist Assistant instance, then the Add New Patient dialog box closes. The new patient records are displayed on the Patients tab. For specific identifying information, asterisks (*) are displayed, and if:

*   You have *not* imported custom fields, then the Add Patient dialog box closes, and a single message, the Patient Import message, opens, indicating the number of patients that were successfully added to the Geneticist Assistant database. Click OK to close the message, and then continue as appropriate in Geneticist Assistant.

Figure 5-7: Patient Import message
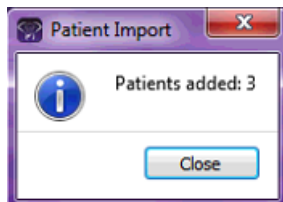
- You have imported custom fields and have selected them for addition to the Geneticist Assistant database, then a first message, a New Custom Fields message, opens indicating that these custom fields will be added to the Geneticist Assistant database.

Figure 5-8:   Custom Fields message, Fields will be added
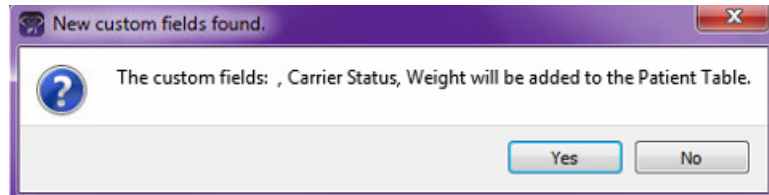
Click Yes to close the New Custom Fields message, and then a second message, a Patient Import message, opens. The message indicates the number of patients that were successfully added to the Geneticist Assistant database opens. Click OK to close the Patient Import message, and then continue as appropriate in Geneticist Assistant.

Figure 5-9:   Number of Patients added message
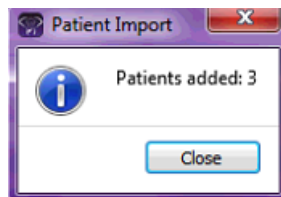
To add patients to the Geneticist Assistant database without encryption

If you do not have encryption enabled for your Geneticist Assistant instance, then an Invalid Encryption Certificate Setting dialog box opens, warning you that no Encryption Certificate is selected and that data will be submitted without encryption, and asking you to continue.

Figure 5-10:   Invalid Encryption Certificate Setting dialog box
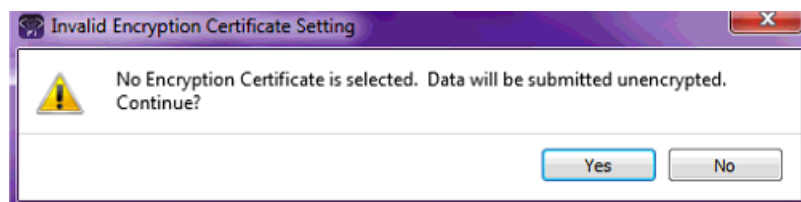
Click Yes.

- If you have *not* imported any custom fields, then both the Invalid Encryption Certificate Setting dialog box and the Add New Patient dialog box close. A single message, the Patient Import message, opens, indicating the number of patients that were successfully added to the Geneticist Assistant database. (See Figure 5-11 on page 134.) Click OK to close the message. The new patient records are displayed on the Patients tab, with all identifying information fully visible.

Figure 5-11:   Number of Patients added message
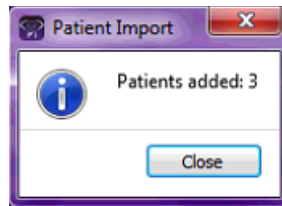
Figure 5-12:   Custom Fields message, Fields will be added
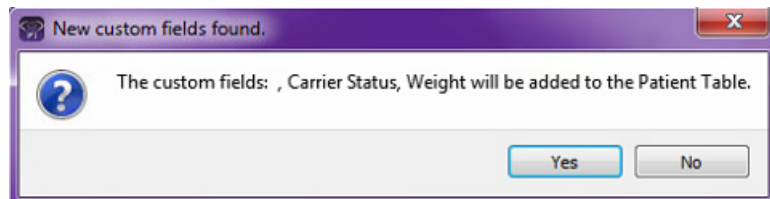
- If you have imported custom fields and have selected them for addition to the Geneticist Assistant database, then the Invalid Encryption Certificate Setting dialog box closes and a first message, a New Custom Fields message, opens indicating that these custom fields will be added to the Geneticist Assistant database.

Click Yes to close the New Custom Fields message, and then a second message, a Patient Import message, opens. The message indicates the number of patients that were successfully added to the Geneticist Assistant database opens. Click OK to close the Patient Import message. The new patient records are displayed on the Patients tab, with all identifying information fully visible. Continue as appropriate in Geneticist Assistant

Figure 5-13:   Number of Patients added message
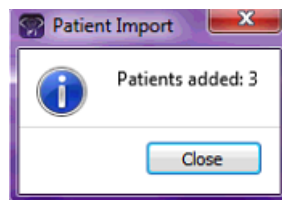
To import a Submissions batch file

You use a batch import process to add multiple patients in a single step to the Geneticist Assistant Customer Portal. 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 in the order listed—the patient ID, the patient last name, the patient first name, the patient date of birth, the patient gender, the patient race, the patient phenotypes, and the patient relationships. Patient relationships must in the format patient ID-Relationship, for example, ABS17-Father. Multiple phenotypes and multiple relationships must be separated by semi-colons. 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.

Figure 5-14: Standard column order and names

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

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

2. Click the Import Submissions tab to open it.

Figure 5-15: Add New Patient dialog box, Import Submissions tab
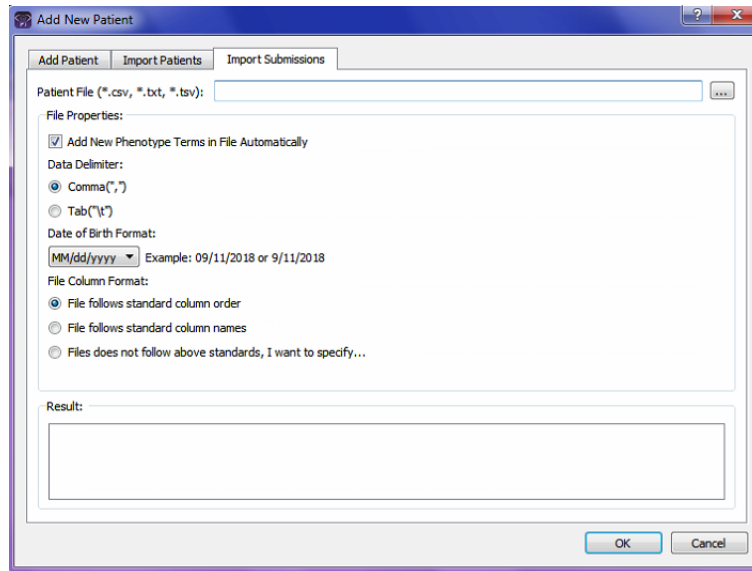
3. Specify the properties of your Submissions file.

Property	Description
Add New Phenotype Terms in File Automatically	Selected by default. If the patient file contains phenotypes that have not already been added to the Geneticist Assistant database either through the Manage Objects function (see "Managing Objects in the Geneticist Assistant Database" on page 55), or the Add Phenotype option on the Add Patient tab (see "Patient tab, Patient sub-tab, Phenotype pane" on page 148), then leave this option selected to automatically add these phenotypes to the database. Otherwise, clear this option.
Data Delimiter	Indicate the delimiter used (Comma or Tab).
Date of Birth Format	Select the format used for the patients' birthdays.

Property	Description
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.

4. If your file follows the standard column order or the standard column names, go to Step 7; otherwise, if your file does *not* follow either of these standards, go to Step 5.

5. 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 "To rearrange the order of data columns" on page 44.*

6. 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 7.

    •   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 7.

7. Click OK.

    The Add New Patient dialog box closes. The patients, and if applicable, any new phenotypes, are imported in to the Customer Portal.

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 editing information (such as adding comments relationships) for existing patients and related patients, and generating, and optionally, saving Patient reports to the Geneticist Assistant database. You can work with patient information from the Patients tab or from the Patient tab.

To review existing patient information on the Patients tab

The Patients tab has a single pane, the Patients pane, for displaying information for existing patients. When you review the information for an existing patient, a variety of options are available for working with the patient, such as editing the identifying information for a patient, and adding relationships and comments for the patient.

1. On the Geneticist Assistant main menu, click Tabs > 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. The patients are sorted in ascending order based on Patient ID. The sorting reflects the order in which the patients were added to the Geneticist Assistant database.

   • 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. The patients are sorted in ascending order based on Patient ID. The sorting reflects the order in which the patients were added to the Geneticist Assistant database.

Figure 5-16:   Patients tab

2. Do any or all of the following as needed for a patient on the Patients tab:

- Edit the identifying information and/or comment for a patient. See "To edit the identifying information and/or comment for a patient" below.

- Right-click a patient on the tab to open a context menu with options for working with the patient. See "To use the context menu for a patient (Patients tab)" on page 141.

- Open the corresponding Patient tab. See "To review existing patient information on the Patient tab" on page 145.
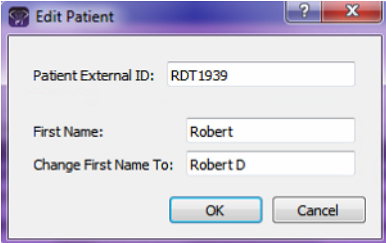
To edit the identifying information and/or comment for a patient
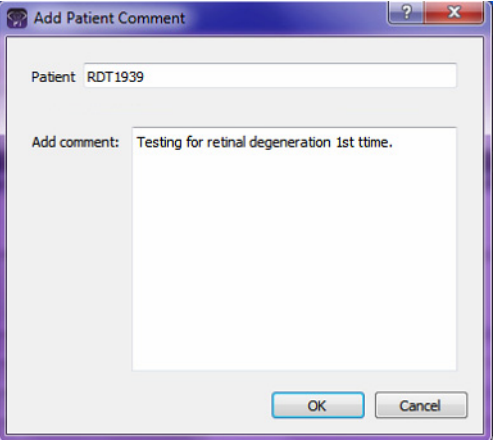
You can edit the identifying information and/or a comment for a patient on the Patients tab.

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

2. For the appropriate patient, do any or all of the following:

Action	Steps
Edit patient gender	1. On the Gender dropdown list, select the appropriate gender. The Edit Patient dialog box opens. The dialog box displays the Patient External ID, the current value for the gender, if applicable, and the new value. *Figure 5-17: Edit Patient dialog box (Gender)* 2. Click OK. The Edit Patient dialog box closes. The Patients tab remains open. The patient record is displayed with the new information.

Action	Steps
Edit patient ethnicity	1. On the Ethnicity dropdown list, select the appropriate ethnicity. The Edit Patient dialog box opens. The dialog box displays the Patient External ID, the current value for the ethnicity, if applicable, and the new value. *Figure 5-18: Edit Patient dialog box (Ethnicity)* 2. If needed, make any other changes to the new value, and then click OK. The Edit Patient dialog box closes. The Patients tab remains open. The patient record is displayed with the new information. **Note:** If the appropriate ethnicity is not available, you can add it. See "Managing Objects in the Geneticist Assistant Database" on page 55.After you add the new ethnicity, you must click Refresh on the Patients tab to make the value available for selection.

Action	Steps
Edit patient identifying information	1. In any of the identifying information fields (External ID, First Name, Last Name, or Date of Birth), single-click twice, and then add or edit the information as needed. **Note:** 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. 2. Click anywhere on the tab that is outside the edited field. The Edit Patient dialog box opens. For any value other than External ID, the dialog box displays the Patient External ID, the current value for the selected information, if applicable, and the new value; otherwise, the dialog box displays only the current Patient External ID, and if applicable, the new value. *Figure 5-19: Edit Patient dialog box (Identifying information)* 3. If needed, make any other changes to the new value, and then click OK. The Edit Patient dialog box closes. The Patients tab remains open. The patient record is displayed with the new information.

Action	Steps
Edit patient comments	1. In the Last Comment field, single-click twice, and then add or edit the comment as needed. 2. Click anywhere on the tab that is outside the edited field. The Add Patient Comment dialog box opens. The dialog box displays the comment. *Figure 5-20: Add Patient Comment dialog box* 3. If needed, make any other changes to the comment, and then click OK. The Edit Patient dialog box closes. The Patients tab remains open. The patient record is displayed with the new information.

To use the context menu for a patient (Patients tab)

You can right-click any patient on the tab to open a context menu with the following options for reviewing and working with the patient record:

Table 5-1:   Patients tab context menu

Option	Description
Patient Detail	Opens the Patient tab. **Note:** You can also double-click a patient to open the Patient tab. See "To review existing patient information on the Patient tab" on page 145.
Edit Patient	Opens the Edit Patient dialog box. You can edit the custom information (fields) for the patient. You can also add new custom fields for the patient. See "To add or edit custom fields on a per patient basis" on page 143.
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 one patient at a time" on page 127 as many times as needed to add all the necessary patient relationships to the patient.
Add All to Custom Report	Selects all the data that is displayed for the patients for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Delete Patient	Deletes the selected patient from the Geneticist Assistant database.

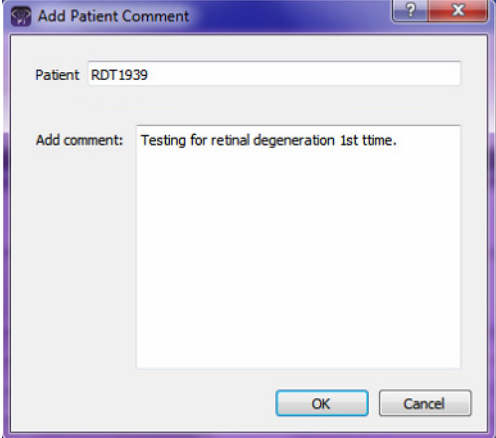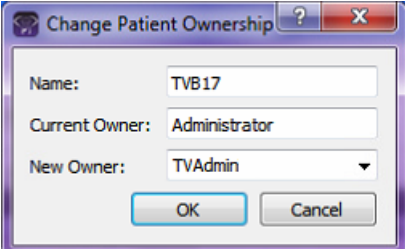*Table 5-1:   Patients tab context menu (Continued)*
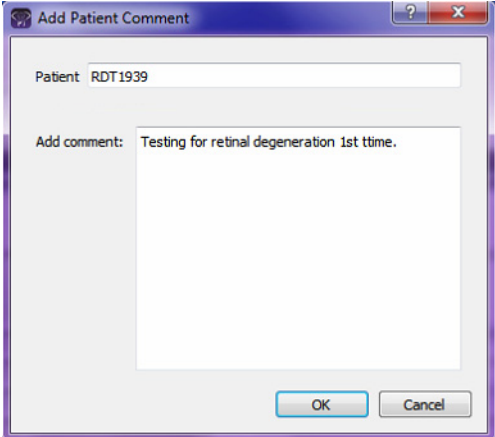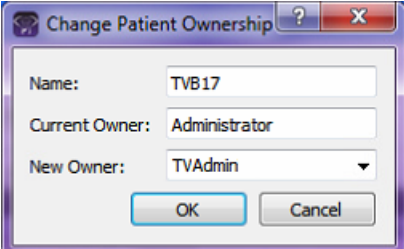
Option	Description
Add Comment	Opens the Add Patient Comment dialog box. You add a comment for the selected patient in the Add Comment field. The comment is displayed in the Last Comment field and in the Comments pane on the Patient sub-tab on the Patient tab. See "Patient tab, Patient sub-tab, Comments pane" on page 152. *Figure 5-21: 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 on the Patients tab. All comments that have been added for the patient are displayed in reverse chronological order in the Comments pane on the Patient tab.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. See "Setting Read and Write Permissions" on page 326.
Change Ownership	Opens the Change Patient Ownership dialog box, which displays the name of the current owner for the patient, and an option (New Owner) for selecting a new owner for the patient from a list of all the currently available and active users for your Geneticist Assistant instance. If you are an Administrator user, the current owner of the patient, or if you have been granted Can Read and Can Write permissions for the patient, then you can the ownership for the patient. *Figure 5-22: Change Patient Ownership dialog box*
Copy	Copies all the information that is displayed for the selected patient to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.

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

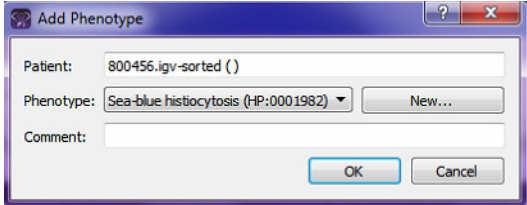| Option	Description
Load Filters	
Manually	Opens the Filters table for the Patients pane. See "To filter the data that is displayed in a tab pane" on page 32.
\<Filter Template Name\> - Saved by \<User Name\>	Opens the saved Filter template for the Patients pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
\<Saved Pane Configuration Name\> - Saved by \<User Name\>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.

To add or edit custom fields on a per patient basis

You use the Import Patients process to batch import patients in to the Geneticist Assistant database. If the batch import file has custom fields in it and you opt to add these fields to the database, then all the same custom fields are imported for all the patients in the batch file. To add or edit custom fields on a per patient basis, you must edit the patient from the Patients tab.
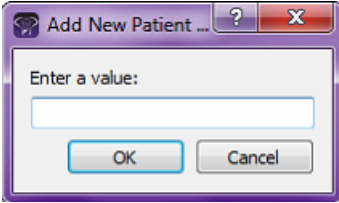
1. If you have not already done so, open the Patients tab.

2. Right-click any selected patient, and on the context menu that opens, click Edit Patient.

   The Edit Patient dialog box opens. The top pane of the dialog displays the read-only identifying information for the patient and the Custom Fields (bottom) pane displays all the custom information (fields) for the patient and their values. See Figure 5-23 on page 144.

Figure 5-23:   Edit Patient dialog box
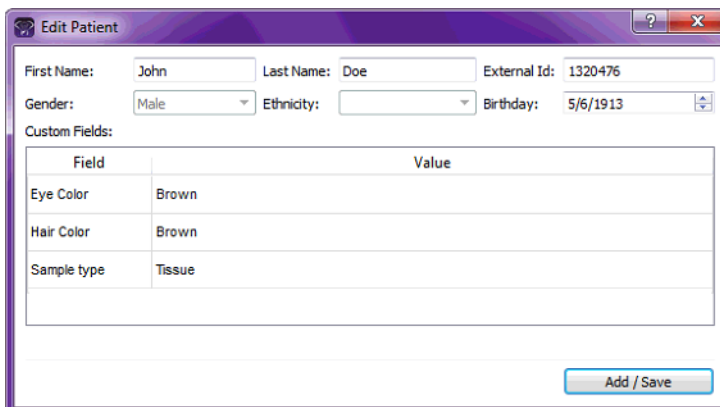
3. Do any of the following as appropriate:

Option	Description
To edit a custom field value	1. Double-click a value to select it, and then edit it as necessary. 2. Click Add/Save.
To clear a custom field value	1. Right-click the value, and on the context menu that opens, click Clear. The current value is deleted.* 2. Click Add/Save.
Note:	*If you have imported custom field in to the Geneticist Assistant database through a Patients batch file, then you can leave the value field blank. If you have added a custom field to a patient through the Edit Patient dialog box, then you cannot leave the value field blank. After clearing the field, you must enter another value; otherwise, if you leave the value field blank, and then save the edited patient, when you open the Edit Patient dialog box again for the patient, the custom field is no longer displayed in the Custom Fields pane.
To add a custom field and value	1. Right-click anywhere in the Custom Fields pane, and on the context menu that opens, click New. An Add New Patient Field dialog box opens. 2. Enter the new custom field name and then click OK. The Add New Patient Field dialog box closes, and the field without a value is displayed in the Custom Fields pane. *Figure 5-24: Add New Patient Field dialog box* 3. Double-click in the Value field for the new custom field, and then enter its value. 4. Click Add/Save.

To review existing patient information on the Patient tab

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

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

2. 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.

3. Continue to one of the following:

    •   "Patient tab, Patient sub-tab" below.

    •   "Patient tab, Samples sub-tab" on page 155.

    •   "Patient tab, Variants sub-tab" on page 158.

Patient tab, Patient sub-tab

When a Patient tab first opens, the Patient sub-tab is the active tab.

Figure 5-25:   Patient tab, Patient sub-tab
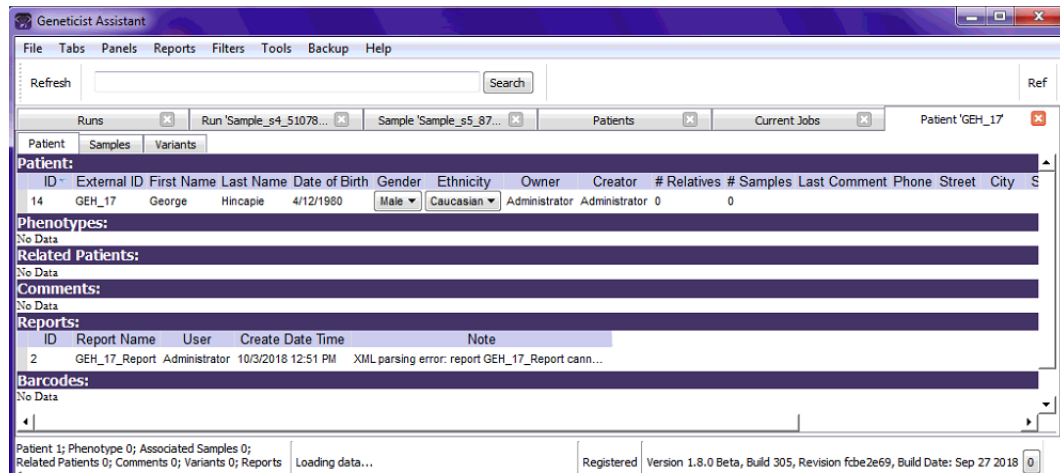
The Patient sub-tab has six panes for displaying and working with patient information.

Pane	Available Options
Patient	• Adding relationships to the patient. • Creating a report for the patient. • Adding comments for the patient. • Settings permissions for the patient. • Specifying the ownership for the patient. See "Patient tab, Patient sub-tab, Patient pane" on page 146.

Pane	Available Options
Phenotypes	Adding and removing phenotypes for the patient. See "Patient tab, Patient sub-tab, Phenotype pane" on page 148.
Related Patients	• Adding relationships to a related patient. • Removing relationships for a related patient. • Adding comments for the related patient. • Settings permissions for the related patient. • Specifying the ownership for the related patient. See "Patient tab, Patient sub-tab, Related Patients pane" on page 150.
Comments	• Adding comments for a patient. • Deleting comments for a patient. See "Patient tab, Patient sub-tab, Comments pane" on page 152.
Reports	• Viewing a Patient report that was saved to the Geneticist Assistant database. • Deleting a Patient report that was saved to the Geneticist Assistant database. See "Patient tab, Patient sub-tab, Reports pane" on page 154.
Barcodes	Viewing all the barcodes that are associated with the patient in the Geneticist Assistant customer portal. See "Patient tab, Patient sub-tab, Barcodes pane" on page 155.

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 edit any of the identifying information for the patient. (Begin at Step 2 of "To edit the identifying information and/or comment for a patient" on page 138.) You can also right-click any patient record in the pane to open a context menu with the following options:

Table 5-1:   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 one patient at a time" on page 127 as many times as needed to add all the necessary patient relationships to 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 10, "Geneticist Assistant Reports," on page 265.

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

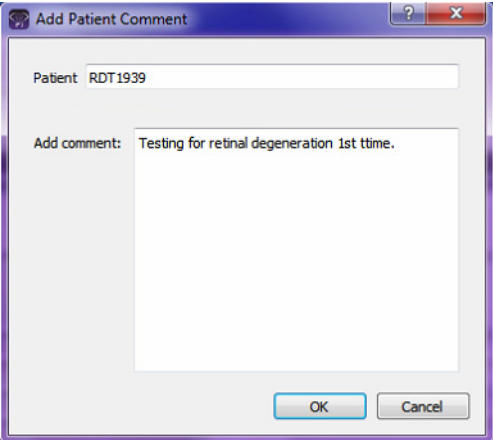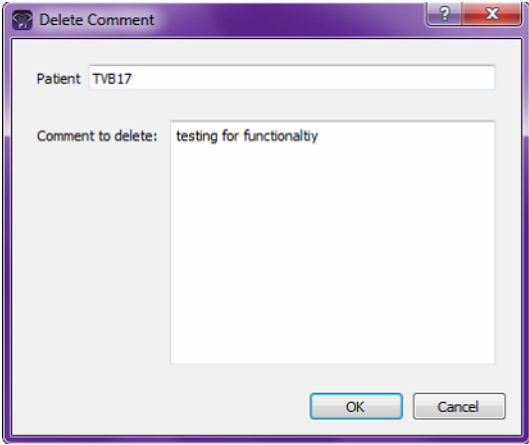| Option	Description
Add Comment	Opens the Add Comment dialog box. You add a comment for the selected patient in the Add Comment field. The comment is displayed in the Last Comment field in the pane and in the Comments pane. See "Patient tab, Patient sub-tab, Comments pane" on page 152. *Figure 5-26: Add 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 edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record.. See "Setting Read and Write Permissions" on page 326.
Change Ownership	Opens the Change Patient Ownership dialog box, which displays the name of the current owner for the patient, and an option (New Owner) for selecting a new owner for the patient from a list of all the currently available and active users for your Geneticist Assistant instance. You can change the ownership for any patient if you are an Administrator user, the current owner of the patient, or if you have been granted Can Read and Can Write permissions for the patient. *Figure 5-27: Change Patient Ownership dialog box*
Copy	Copies all the information that is displayed in the Patient pane for the patient to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.

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

Option	Description
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
\<Saved Pane Configuration Name\> - Saved by \<User Name\>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Patient pane. See "To filter the data that is displayed in a tab pane" on page 32.
\<Filter Template Name\> - Saved by \<User Name\>	Opens the saved Filter template for the Patient pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Patient tab, Patient sub-tab, Phenotype pane

The Phenotype pane on the Patient sub-tab shows all the phenotypes that have been added for the patient, and if applicable, the associated comments that have been added for each phenotype. You can right-click in the pane to open a context menu with the following options:

Table 5-2:   Patient tab, Patient sub-tab, Phenotypes pane context menu
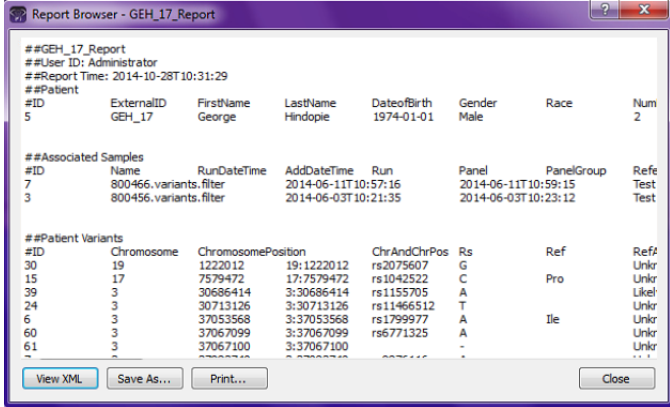
Option	Description
Add All to Custom Report	Selects all the data that is displayed in the Phenotype pane for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.

Table 5-2:   Patient tab, Patient sub-tab, Phenotypes pane context menu (Continued)

Option	Description
Add Phenotype	Opens the Add Phenotype dialog box. The Phenotypes dropdown list contains all the phenotypes that are currently available in the Geneticist Assistant database that you can add for the patient. You can add multiple phenotypes for the patient. You can also add a comment for each phenotype that you add for the patient. *Figure 5-28: Add Phenotype dialog box* **Note:** If the appropriate phenotype is not available, then you can click New to add it to the Geneticist Assistant database. You can also use the Manage Objects function to add a phenotype to the Geneticist Assistant database. See "Managing Objects in the Geneticist Assistant Database" on page 55.
Delete Phenotype	Deletes the selected phenotype from the patient record. The phenotype is not deleted from the Geneticist Assistant database.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Phenotypes pan. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Phenotypes pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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 pane 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 also right-click in the Related Patients pane to open a context menu with the following options:

Table 5-3:   Patient tab, Patient sub-tab, Related Patients pane context menu
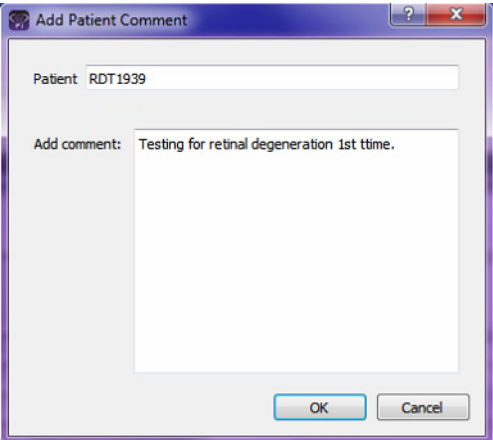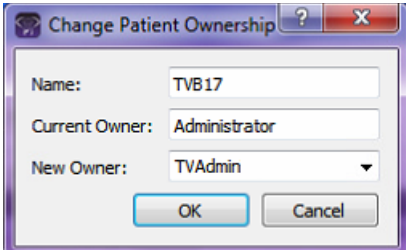
Option	Description
Reload in Current Tab	Displays the information for the selected related patient in the currently opened Patient tab.
Open in New Tab	Displays the information for the selected related patient in a new Patient tab. The currently opened Patient tab remains open, but the focus is changed 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 one patient at a time" on page 127 as many times as needed to add all the necessary patient relationships to the related patient.
Remove Selected Relationship	Removes the selected relationship 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 10, "Geneticist Assistant Reports," on page 265.
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 5-29:   Add 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 5-3: 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 edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. See "Setting Read and Write Permissions" on page 326.
Change Ownership	Opens the Change Patient Ownership dialog box, which displays the name of the current owner for the related patient, and an option (New Owner) for selecting a new owner for the related patient from a list of all the currently available and active users for your Geneticist Assistant instance. You can change the ownership for any patient if you are an Administrator user, the current owner of the patient, or if you have been granted Can Read and Can Write permissions for the patient. *Figure 5-30: Change Patient Ownership dialog box*
Copy	Copies all the information that is displayed for the related patient to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Related Patients pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Related Patients pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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. If one or more comments are displayed in the pane, then you can right-click in the pane to open a context menu with the following options:

The context menu for the Comments pane is not enabled if the pane is blank. To a comment for the patient and enable this context menu, see "To use the context menu for a patient (Patients tab)" on page 141.

Table 5-4:   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 5-31:   Add Comment dialog box

Table 5-4:   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 patient and close the dialog box. *Figure 5-32: Delete Comment dialog box*
Copy	Copies all the information that is displayed for the selected comment to your client's clipboard. You can then use the standard Paste commands to paste this copied information into a third-party application such as Notepad.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Comments pan. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Comments pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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 includes these same reports.

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

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

Option	Description
View Report	Opens the Report Browser dialog box, which displays the report in plain text. **Note:** You can also double-click the report entry to open the Report Browser dialog box. *Figure 5-33: 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	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.

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

Option	Description
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Reports pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Reports pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

> *For detailed information about generating and saving both standard and custom Patient reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.*

Patient tab, Patient sub-tab, Barcodes pane

The Barcodes pane on the Patient sub-tab lists all the barcodes that are associated with the patient in the Geneticist Assistant customer portal. You can use these barcodes to validate that the correct patient/sample combination was submitted to the customer portal and to confirm sample identity when you are reviewing a sample in Geneticist Assistant. See Chapter 12, "The Geneticist Assistant Customer Portal and Submissions," on page 305.

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 samples.

Figure 5-34:   Patient tab, Samples sub-tab
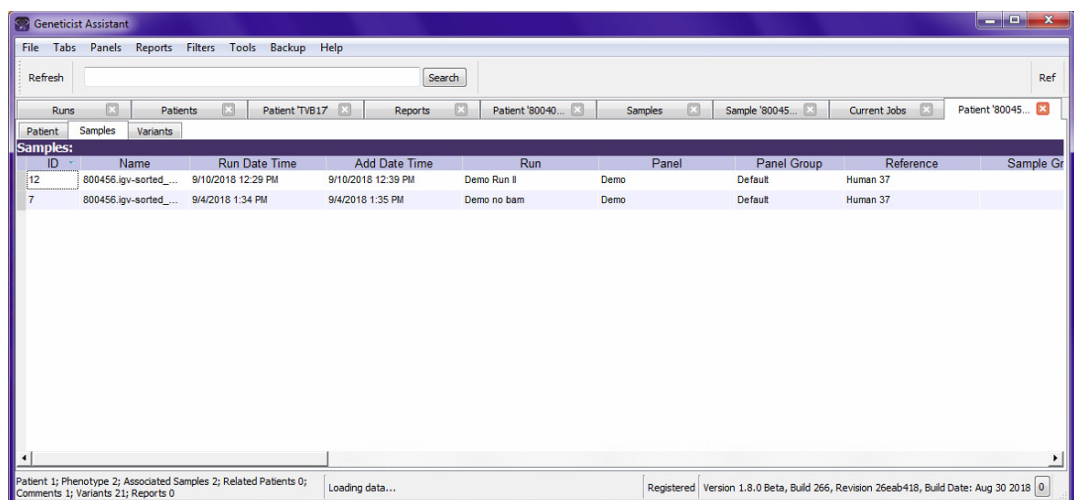
You can do the following on the Samples sub-tab:

- View the details for an individual patient sample.

- View the sample variants filtered by a selected panel.

- View the coverage information for all regions in a sample panel.

- Calculate the panel statistics for a patient sample.

- Set sample permissions.

- View the patient samples in a third-party viewer (NextGENe or IGV).

- Export the patient samples to a VCF file.

- Generate Sample reports for one or more selected samples.

- Generate a Batch Variants report for a selected sample or samples.

- Generate a Batch Coverage report for a selected sample or samples.

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

Table 5-6: Patient tab, Samples sub-tab, Samples pane context menu
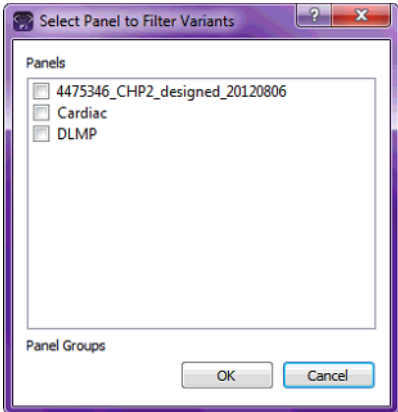
Option	Description
Sample Details	Opens the Sample tab, which contains detailed information about the associated sample, including the variant list. See Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209. **Note:** You can also double-click the sample to open the Sample tab.
Show Sample Variants Filtered by Panel	Opens the Select Panel to Filter Variants dialog box. Select a panel in the dialog box to show the variants in the patient's sample filtered by the selected panel or panels. *Figure 5-35: Select Panel to Filter Variants dialog box*
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 tab" on page 200.
Panel Statistics	Opens the Panel Statistics tab. The tab has options for displaying specific information based on the panel that was used for the sample. See "Reviewing Panel Statistics" on page 319.

Table 5-6:   Patient tab, Samples sub-tab, Samples pane context menu (Continued)

Option	Description
Copy	Copies all the information that is displayed for the selected associated sample to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.
Permissions	Opens the Permissions dialog box. You use the options on this dialog box to set or edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient sample. See "Setting Read and Write Permissions" on page 326.
External - Contains all the sub-menu options for viewing the sample variants in a tool that is outside Geneticist Assistant.	
View in NextGENe	Requires you to have your NextGENe installation specified in your Geneticist Assistant settings. (See "To configure Geneticist Assistant directories" on page 59.) If selected, then Geneticist Assistant browses for the folder where the VCF file for the sample is located. If the NextGENe project file is found in the same directory as the VCF file, then the NextGENe project is opened in the NextGENe Viewer.
View in IGV	Applicable if there is a corresponding BAM file (and not a NextGENe project file) in the same folder as the VCF file for the sample. Opens the BAM file in the third-party Integrative Genomics Viewer (IGV).
Export - Contains all the sub-menu options for exporting a variant.	
Export to VCF	Exports all the variants in the selected sample or samples (CTRL-click to select multiple samples) to a VCF file. You must specify both the file name and the location in which to save the file.
Save Sample Reports	Applicable for one or more samples. CTRL-click to select multiple samples. Generates an individual sample report for each selected sample. If a single sample is selected, then a default location, report name, and report type (.csv) are provided, but you can change any or all of these values. If multiple samples are selected, then you can select only the location in which to save the report. Each report is named the same as the sample and the report format is .csv, and you cannot change either of these values. **Note:** For more information about standard reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.
Create Batch Variants Report	Generates the Batch Variants report for the selected sample or samples. (CTRL-click to select multiple samples.) See "Managing a Batch Variants Report" on page 329.
Create Batch Coverage Report	Generates the Batch Coverage report for the selected sample or samples. (CTRL-click to select multiple samples.) See "Generating a Batch Coverage Report" on page 335.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.

Table 5-6: Patient tab, Samples sub-tab, Samples pane context menu (Continued)

Option	Description
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Samples pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Samples pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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 available in the Variants pane is identical to the information that is available for a variant on the Sample Variant Data sub-tab on the Sample tab. (See "Sample Variant Data Sub-Tab" on page 212.)

Figure 5-36: Patient tab, Variants sub-tab
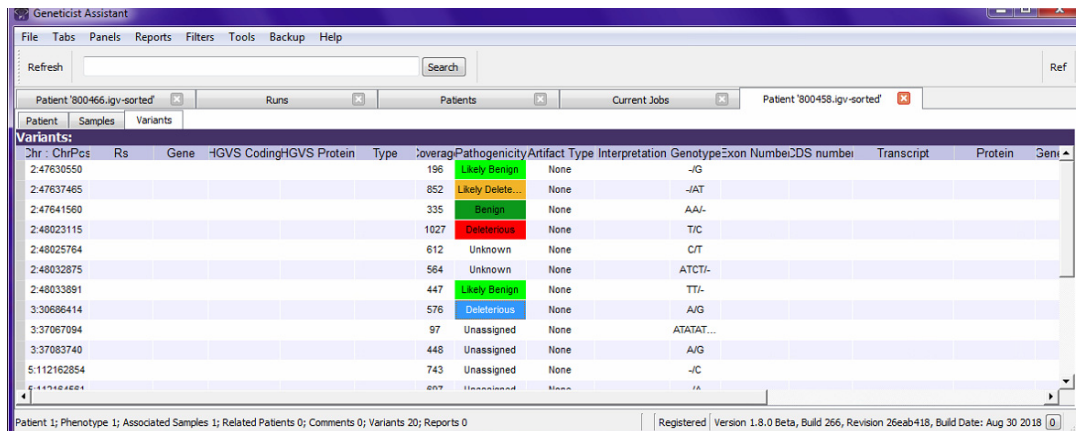
You can specify how the information about the variants is to displayed on the Variants sub-tab. See "To specify variant preferences" on page 70. You can also do the following for an individual variant that is displayed on the Variants sub-tab:

- Select a different gene for a variant: If the variant is located where there are overlapping genes, then you can select a different gene for the variant on the Gene dropdown list. If you select a different gene, then the gene that is used on the current tab for the current Geneticist Assistant session is temporarily changed. After you close and then re-open Geneticist Assistant, the selected gene is reset to the preferred gene. To permanently change the gene that is used for variant, you can add a preferred gene for the panel. See

"To set or delete a preferred gene for a panel" on page 112.

- Select a different transcript for a variant: You can select a different transcript for the variant on the Transcript dropdown list. If you select a different transcript, then the transcript that is used on the current tab for the current Geneticist Assistant session is temporarily changed. After you close and then re-open Geneticist Assistant, the selected transcript is reset to the preferred transcript.

To permanently change the transcript that is used for a gene or genes, you can do one of two things: You can right-click the variant, and on the context menu that opens, click Edit Variant > Update Preferred Transcript (see "Patient tab, Variants sub-tab context menu" on page 160), or you can add a preferred transcript list for the panel. See "To set or delete a preferred transcript list for a panel" on page 110.

- Add or edit a comment for a variant: 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 Add Variant Comment dialog box opens.

Figure 5-37: Add Variant Comment dialog box
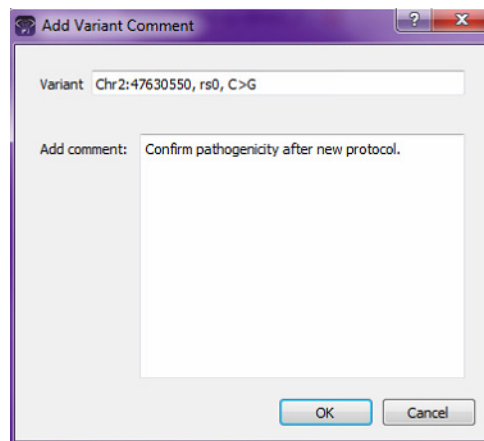
The dialog box displays the variant for which you are adding or editing the comment, and if applicable, any current comment. You can add a new comment or make edits to the current comment, and then click OK to close the dialog box and save the new or modified comment for the variant.

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.

- Edit the type and/or the HGVS information (Genomic, Coding, and/or Protein) for a variant: You can click once in the Type field, the HGVS Genomic field, the HGVS Coding field, or the HGVS Protein field to select the current value in the field and edit it.

> *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 251. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in Geneticist Assistant.*

- You can single click twice in the Interpretation field to select the current interpretation in the field and edit it, or if the field is blank, enter a new interpretation. After you add or edit an interpretation and then click out of the field, the Add Variant Interpretation dialog box opens.

Figure 5-38: Add Variant Comment dialog box
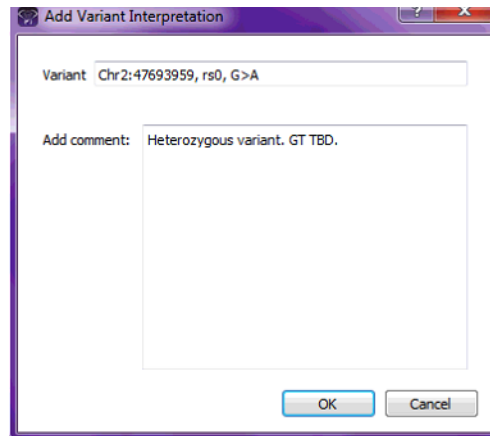
The dialog box displays the variant for which you are adding or editing the interpretation and the current interpretation. You can edit the current interpretation, or leave it as-is, and then click OK to close the dialog box and save the variant interpretation.

Patient tab, Variants sub-tab context menu

You can right-click a variant on the Variants sub-tab to open a context menu with the following options:

Table 5-7: Patient tab, Variants sub-tab context menu

Option	Description
Variant Details	Opens the Variant tab for the selected variant. See "Overview of the Variant Tab" on page 245. **Note:** You can also double-click the variant to open the Variant tab.

Table 5-7:   Patient tab, Variants sub-tab context menu (Continued)
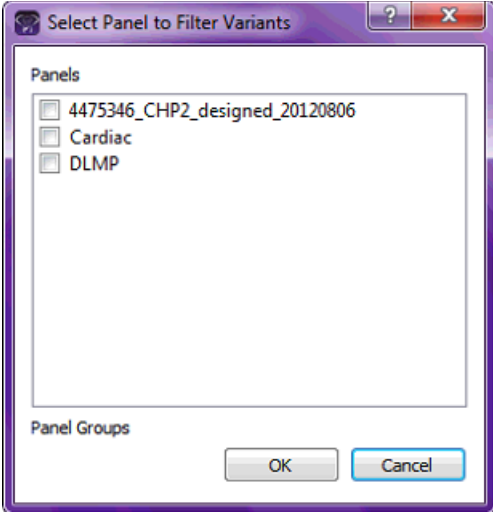
Option	Description
Show Variants Filtered by Panel	Displays all the variants in all the associated samples for the selected patient filtered by a selected panel or panels. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel or panels in this dialog box by which to filter the variants for the selected patient. *Figure 5-39: Select Panel to Filter Variants dialog box*
Show Samples with Selected Variants	
All Variants	Select a variant, or CTRL-click to select multiple variants and then select this option to open the Samples with all variants pane, which displays a list of all the samples in your Geneticist Assistant database that contain all the selected variants. You can right-click a sample in the list to open a context menu with sample-specific options. See "The Samples Pane (Run tab) Context Menu" on page 191.
Any Variants	Select a variant, or CTRL-click to select multiple variants and then select this option to open the Samples with all variants pane, which displays a list of all the samples in your Geneticist Assistant database that contain one or more of the selected variants. You can right-click a sample in the list to open a context menu with sample-specific options. See "The Samples Pane (Run tab) Context Menu" on page 191.

Table 5-7:   Patient tab, Variants sub-tab context menu (Continued)
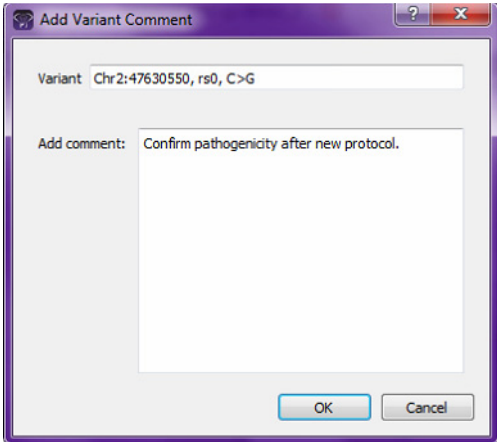
Option	Description
Edit Variant - Contains all the sub-menu options for editing and deleting a variant.	
Add Variant Comment	Opens the Add Variant Comment dialog box. You can add a comment for a selected variant in the Add Comment field, and then click OK. You can also CTRL-click to select multiple variants and add the same comment to all the selected variants in a single step. 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 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 5-40:   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.

<Add Variant Interpretation>	Select a variant, or CTRL-click to select multiple variants. • If a single variant is selected, then the option is Add Single Variant Interpretation, and the Add Single Variant Interpretation dialog box opens. • If two or more variants are selected, then the option is Add Compound Variant Interpretation and the Add Compound Variant Interpretation dialog box opens. See "Adding a Variant Interpretation" on page 342.

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

Option	Description
Update 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, then you can set the status to Unknown.) When you update the pathogenicity and/or status, you can apply the changes to: • the panel group - Applies the changes to the variant for the indicated panel group.The next time that the variant is detected in another sample that has been assigned to this panel group, then this is the pathogenicity that is displayed for the variant. • the sample only - Applies the changes to the variant only for the currently selected sample. You can also update the pathogenicity status and/or add comments for the changes. By default, the status is always set to an initial value of Pending. *Figure 5-41: Submit Pathogenicity dialog box* **Note:** You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box. The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five pathogenicities—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities, either by clicking New to open the Add New Pathogenicity dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. • By default, only two statuses—Pending and Confirmed—are available. You have the option of adding custom statuses, either by clicking New to open the Add New Pathogenicity Status dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. • Optionally, you can add supporting information for this pathogenicity change. See "To add supporting information for a variant" on page 256. • You always have the option of updating just the pathogenicity status for a variant. See "Update Pathogenicity Status" on page 164. • You can also update the pathogenicity on the Sample tab, Sample Variant Data sub-tab. See "Sample Variant Data Sub-Tab" on page 212.

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

Option	Description
Update Pathogenicity Status	Select a variant, or CTRL-click to select multiple variants and then select this option to open the Submit Pathogenicity Status dialog box. You can update the pathogenicity status for the selected variants in this dialog box. When you update the pathogenicity status, you can apply the changes to: • the panel group - Applies the changes to the variant for the indicated panel group. The next time that the variant is detected in another sample that has been assigned to this panel group, then this is the pathogenicity status that is displayed for the variant. • the sample only - Applies the changes to the variant only for the currently selected sample. • You can also add a comment for the status change. By default, the pathogenicity status is always set to an initial value of Pending. You must review the information for the variant and then decide what the pathogenicity status should be. By default, only two statuses—Pending and Confirmed—are available. You have the option of adding custom statuses, either by clicking New to open the Add New Pathogenicity Status dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. *Figure 5-42: Submit 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 also update the Pathogenicity Status for a variant on the Sample tab, Variant Data sub-tab. See "Sample Variant Data Sub-Tab" on page 212.

Table 5-7:   Patient tab, Variants sub-tab context menu (Continued)
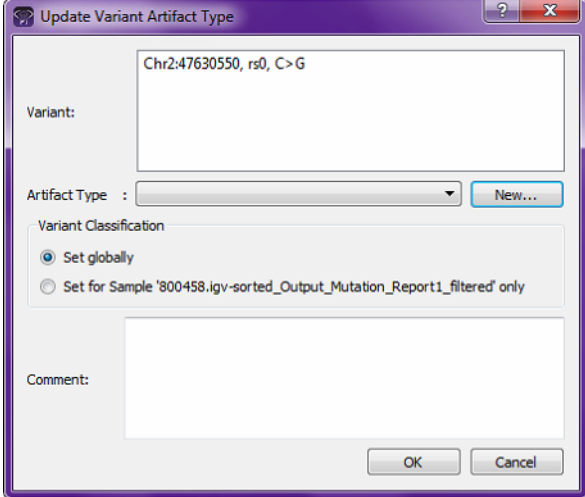
Option	Description
Update Artifact Type	Select a variant, or CTRL-click to select multiple variants and then select this option to open the Update Artifact Type dialog box. You specify the artifact type (for example, Sequencing) for the variant in this dialog box. When you specify the artifact type, you can set the artifact type as the following: • Set globally—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. **Note:** The default value for Variant Classification is determined by the setting on the Variant Preferences tab on the Settings dialog box. See "To specify variant preferences" on page 70. You have the option of adding custom artifact types, either by clicking New to open the Add New Artifact dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. *Figure 5-43: Update Variant Artifact Type dialog box* **Note:** 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 Panel Statistics tab from the Runs tab or the Run tab, or on the Sample Variant Data sub-tab on the Sample tab.

Table 5-7:   Patient tab, Variants sub-tab context menu (Continued)
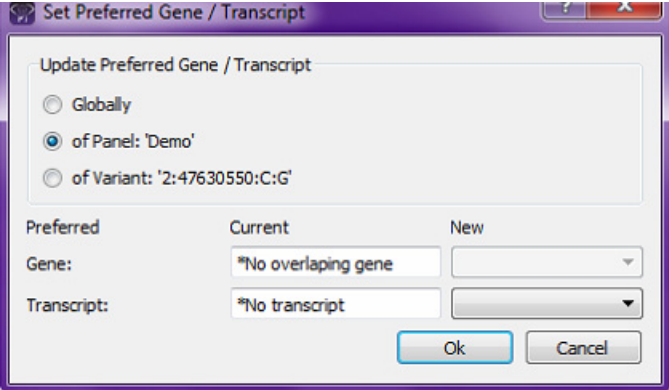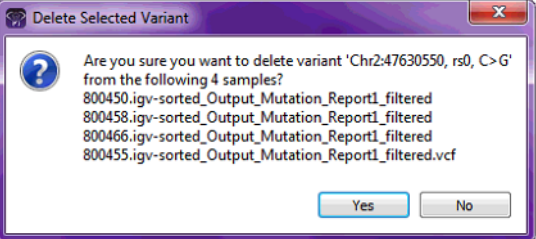
Option	Description
Update Preferred Transcript	Opens the Set Preferred Gene/Transcript dialog box. You can select a different preferred gene and/or transcript for the variant. When you select a different gene/ preferred transcript, you can set the preferences for one of the following: • globally - for all panels and panel groups. • of panel - for only the indicated panel. • of Variant - only for this particular variant and not for any other positions within this panel or panel group. *Figure 5-44: Set Preferred Gene/Transcript dialog box* **Note:** When you select a different transcript here for a panel, then the preferred transcript that is used for a gene or genes is permanently changed for the panel. You can also permanently change the preferred transcript by adding a preferred transcript list for the panel. See "To set or delete a preferred transcript list for a panel" on page 110. To temporarily change the preferred transcript for a gene or genes, you simply select the transcript on the Transcript dropdown list on a tab or sub-tab.
Delete Variant from This Patient	Deletes the selected variant from all samples *for the selected patient* in which it was found. *Figure 5-45: Delete Selected Variant dialog box*

Table 5-7: Patient tab, Variants sub-tab context menu (Continued)
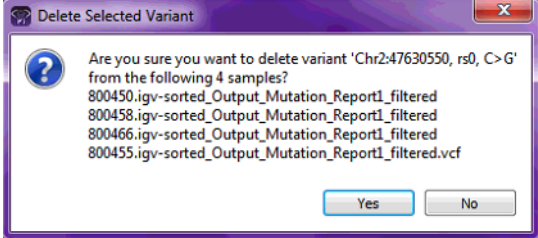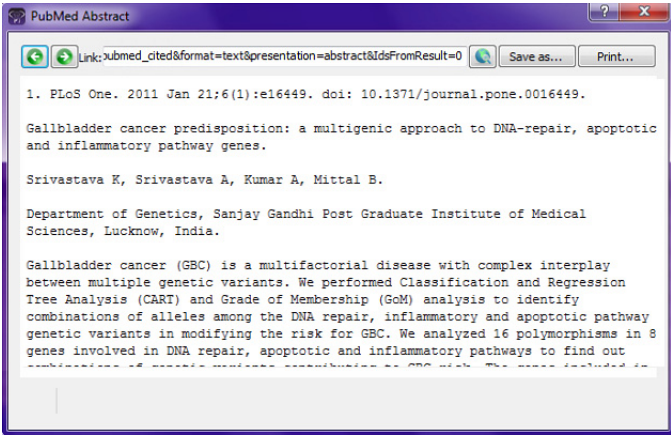
Option	Description
Delete Variant from All Samples	Deletes the selected variant from all samples in which it was found. *Figure 5-46: Delete Selected Variant dialog box*
External - Contains all the sub-menu options for viewing the sample variants in a tool that is outside Geneticist Assistant.	
View in IGV	Opens the variant for viewing in the Integrated Genomics Viewer. **Note:** To view a variant in IGV, your IGV settings must be specified. See "To specify the IGV settings" on page 75.
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 third-party (Alamut, IGV, OMIM) settings" on page 74.
View in UCSC	Opens the variant for viewing in the UCSC Genome Browser.
View PubMed Abstract	Searches PubMed for the variant and displays the appropriate abstract in the PubMed Abstract window. *Figure 5-47: PubMed Abstract 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.

Table 5-7:   Patient tab, Variants sub-tab context menu (Continued)
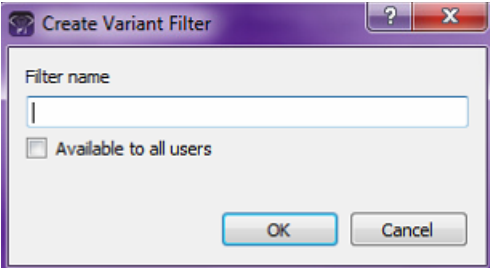
Option	Description
Export - Contains all the sub-menu options for exporting a variant.	
Export Selected Variants to VCF	Exports the selected variants to a VCF file. CTRL-click to select multiple variants. You must specify both the file name and the location in which to save the file.
Export All Variants 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 Custom Report	Selects all the data that is displayed for the selected variant for inclusion in a custom report. CTRL-click to select multiple variants. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Add All Variants to Custom Report	Selects all data that is displayed for all the variants for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Copy	Copies the information for the selected variant to your client's clipboard. You can use standard Paste commands to paste the copied information in to a third-party application such as Notepad.
Create Variant Filter from Selected Variants	Create a filter based on the selected variants. You can select a single variant, or CTRL-click to select multiple variants to open the Create Variant Filter dialog box. Name the filter and indicate if the filter is to be available to all users. You, and if applicable, other users can use the Load Filter Template function to apply this variant filter to a selected sample or samples. (See "To load a saved filter template" on page 38.) *Figure 5-48: Create Variant Filter dialog box* When the Sample tab opens for a sample to which the filter has been applied, then the Sample Variant Data sub-tab displays only those variants that meet the filter criteria.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.

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

Option	Description
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Variants pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Variants pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Adding a Run

A run refers to loading a BAM 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 edit 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 file to include coverage information for the panel regions.

Geneticist Assistant supports only sorted VCF files.

To add a run

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

   The New Run dialog box opens.

Figure 5-49: 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.

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	If the correct references are not available, then you might need to specify the reference directory and/or add the necessary references to the directory. For information about specifying the reference directory, see "Specifying your Geneticist Assistant Settings" on page 59. For information about downloading the most recent references, see "Updating References" on page 48. **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	After you select a panel, the panel group is automatically selected and the name is displayed in the Panel Group field. **Note:** If the correct panel is not available, then you can add it. Click New to open the Manage Panels dialog box and import the needed panel. See "Importing a Panel" on page 105. **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 Group	Read-only field that displays the name of the group to which the selected panel belongs.
Quality Metric Profile	If the correct quality metric profile is not available, then you can add it. Click New to open the Manage Quality Metrics dialog box and add the profile. See "Managing Quality Metrics" on page 77.
Owner	By default, Owner is set to your user name. Optionally, you can select a different owner for the run. If the appropriate user is not available, and you are an Administrator user or a user with the correct permissions, then you can add the new user. Click New to open the Add New User dialog box and add the new user. See "Managing Users" on page 87.
Multiple Samples in VCF	Select this option if you are loading a VCF file that contains variants from multiple samples.
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 a value, select the value, 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 you are changing 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.
User Group	By default, non-administrative users have read/write access to only those runs that they submitted. If other non-administrator users are to have read/write access to this run, then select the appropriate user group.

Setting	Description
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.
Subpanel	To verify coverage for a smaller number of regions that are within the panel that has been selected for the run, select the appropriate subpanel. **Note:** If no subpanels have been created for the selected panel, then no options are available on the Subpanel dropdown list. For information about creating subpanels, see "Adding a Subpanel" on page 119.
Submit Coverage	Selected by default. If BAM files are included in the run, then select this option to import coverage information for the panel regions from the BAM files. Clear this option to skip the coverage import.
Import from VCF	Selected by default. If you are loading a VCF file that contains pathogenicity information (pathogenicity and status) and variant comments, then by default, this information is imported into the Geneticist Assistant database. • Pathogenicity and status - Selected by default. **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. • Comment - Selected by default. • Artifact type - Select to submit any artifact any information that the loaded VCF file contains.
Submit Hotspots	If any variants in the loaded VCF file have an allele frequency value = 0, then select this option to submit these variants to the Geneticist Assistant database.
Accepted VCF filters	Applicable after you load sample files. See Step 5.
Identify Patient ID	
Add New Patient	If the correct patient is not available to associate with a sample for a run, then you can click this option to open the Add New Patient dialog box and add the patient. See Step 6.
• from Sample Name	• Automatically match a patient ID based on the sample file name and the selected separator. For example, if the separator is a period, then all text up to the first period in the sample file name is used as the patient ID.
• from File	• Match the sample with the patient ID as defined in the provided tab-separated values file, with 1 patient/sample per line in the file containing two fields: sample name followed by a tab, and then patient ID.
Split Sample Name by Separator	Selected by default. If you clear this option, then the entire VCF sample file name is used as the patient ID. Select the separator that you are using to define the patient ID from the sample file name.

4. Do one of the following:

- To automatically load all the applicable sample files (VCF only or VCF and BAM files) that are in the same folder in a single step, do the following:

  - Click Select Directory to open the Select Directory dialog box,

  - In the Select Directory dialog box, browse to and select the appropriate directory, and then click Select Folder.

- To manually select sample 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 BAM coverage file 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 or BAM file for a new run that has been submitted previously for another run. You can keep the sample name the same, or you can edit the sample name as needed.

- You can use the same run name to load additional sample files (BAM and/or VCF) 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 file to include coverage information for the panel regions.

- If you have a *single* sample to load, and the VCF and BAM file names do *not* contain corresponding fields in the file names, then select the VCF or BAM 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 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 files do *not* contain corresponding fields in the file names, then CTRL-click as you select only the VCF files or the BAM 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 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 file, and the files *do* contain corresponding fields in the file names, then CTRL-click as you select both the VCF and BAM 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 combination this way, or you can load multiple VCF/BAM file combinations. Each correspondingly named VCF/BAM 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 303.

- 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 edit the sample name.

- If you load a sample file in error, then you can right-click anywhere in the sample entry area, 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.

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

Figure 5-50:   New Run dialog box with loaded sample files
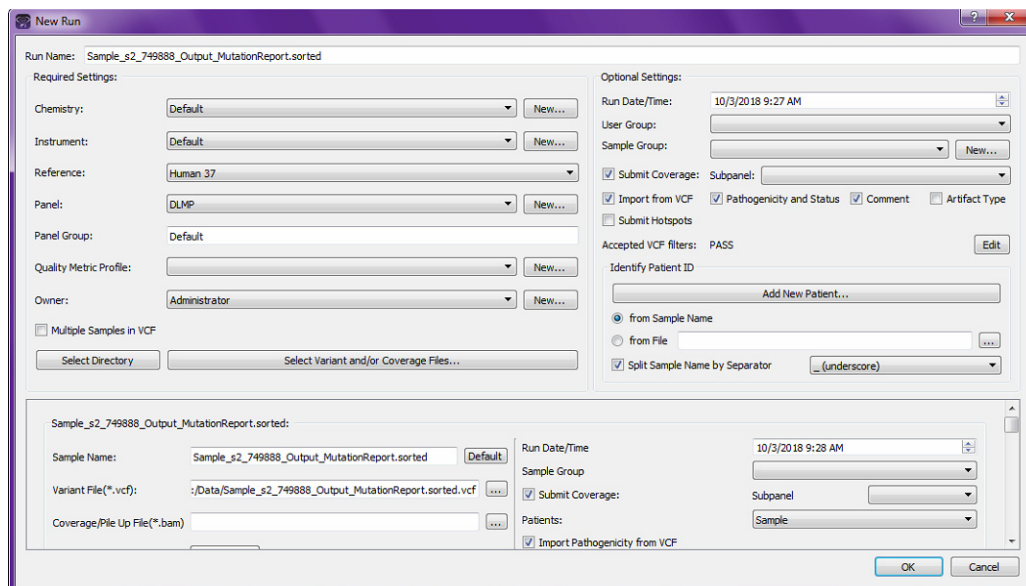
5. After you load your sample VCF files, do one of the following:

    *   To import only those sample variants that have been flagged as passing (Pass) the filter values in the loaded VCF files into your Geneticist Assistant database, leave Accepted VCF filters set to the default value of Pass.

    *   To set additional filter types to be imported from the VCF, click Edit to open the Select Accepted Filters dialog box, and then select the values from the list of available filters. The options that are available depend on the filter values that are included in the loaded VCF files.

Figure 5-51:   Selected Accepted Filters dialog box
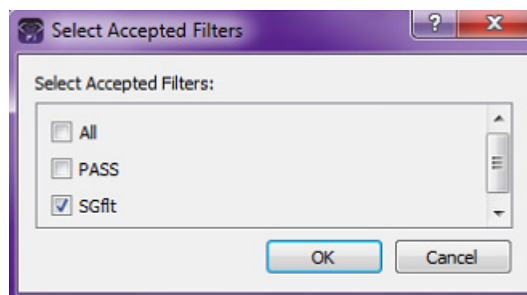
This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.

After you select the filters, and then click OK, the Select Accepted Filters dialog box closes. The selected filters replace the default value of All.

Figure 5-52:   Selected filters specified for a VCF file on the New Run dialog box
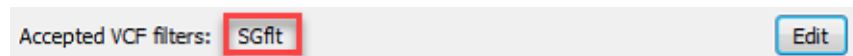
6. Optionally, for each sample, do any or all of the following:

    *   Edit 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 edit any or all these values on a per sample basis.

If you edit the panel on per sample basis, then the panel group might also be modified, but you cannot edit 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 127.

- By default, if you have specified variant filters, then these filters are applied at the run level. To edit the accepted variant filters for a sample, click Edit, and then in the Select Accepted Filters dialog box, edit the filter selection.

7. 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 - Runs Tab," on page 177.

Chapter 6

Geneticist Assistant Data Review - Runs Tab

Many options are available from the Runs tab for reviewing your data in Geneticist Assistant.

This chapter covers the following topics:

- "Overview of the Runs Tab" on page 179.
- "The Runs Tab Context Menu" on page 181.

Overview of 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 the 30 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 the 30 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 also open the Runs tab from the Tabs option on the Geneticist Assistant main menu: Tabs > Runs.

Figure 6-1:   Runs tab
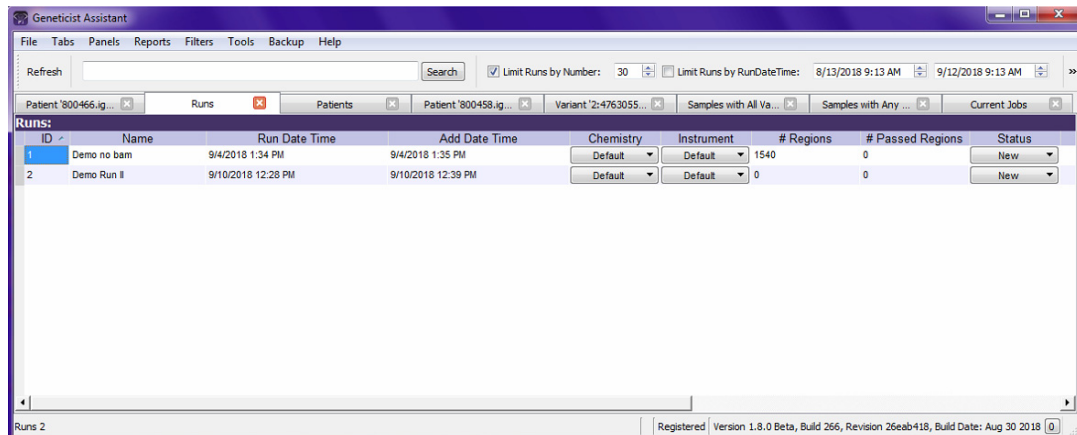
You can do the following on the Runs tab:

- Change the number of runs that are displayed on the tab. See "To change the number of runs displayed on the Runs tab" on page 180.

- Edit certain values that are displayed for a run. See "To edit values for a selected run" on page 180.

- Open a context menu with a variety of options for working wit h and displaying run-specific data. See "The Runs Tab Context Menu" on page 181.

To change the number of runs displayed on the Runs tab

You can do the following for the display on the Runs tab:

- Limit the runs that are displayed by number. The default value is 30.

- 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.

To edit values for a selected run

For any run that is displayed on the Runs tab, you can:

- Click in any of the following fields—Name or Run Date/Time—to select the current value, and edit it as necessary, and then click out of the field.

- Select a different Chemistry and/or Instrument for the run.

  After you edit 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.

Figure 6-2:   Edit Run dialog box
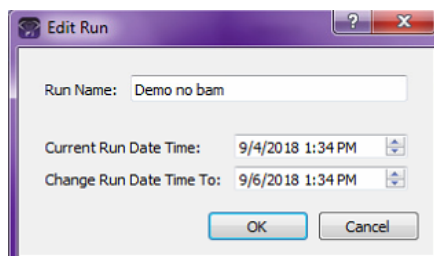
- Set the status for the run. When a run is first submitted, the status is set to New. You can change the status from New to one of three default values (QC Passed, Reviewed, or Complete) for the run, or if custom values have been added for the run, then you can also select one of these values.

For information about adding a custom value, see "Managing Objects in the Geneticist Assistant Database" on page 55.

The Runs Tab Context Menu

You can right-click any run entry on the Runs tab to open a context menu with the following options:

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

Option	Description
Run	• Open – Opens the Run tab for the selected run. See Chapter 7, "Geneticist Assistant Data Review - Run Tab," on page 187. **Note:** You can also double-click a run entry to open the Run tab. • Open all Samples with <Filter>: Select a filter for the samples. One or more Sample tabs open. The Sample Variant Data sub-tab is the open tab and the selected filter is applied to each tab. For each sample, only those sample variants that meet the filter criteria are displayed on the tab. The selected filter is persistent within this Geneticist Assistant instance and across instances. To clear the filter only for the current instance of Geneticist Assistant, click Clear at the bottom of the Table Filters pane. To select a different filter for the samples, or to permanently clear the filter for the samples, see "Apply Variant Filter" under "Edit Sample" on page 193. **Note:** For information about creating a filter template, see "To filter the data that is displayed in a tab pane" on page 32. For information about the Sample Variant Data sub-tab, see "Sample Variant Data Sub-Tab" on page 212.
Show Run Variants • All • Distinct • All Filtered by Panel • Distinct Filtered by Panel	Displayed for one or more runs. CTRL-click to select multiple runs. You can select only one of the four options. All options open the Sample Variant Data sub-tab for the Sample tab, which displays variants for one or more samples. • Displays all the variants for all the samples in all the selected runs. • Displays all the variants that are different among all the samples in all the selected runs. • Displays all the variants among all the samples in all the selected runs filtered by the selected panel. • Displays all the variants that are different among the selected samples filtered by the selected panel.
Add All to Custom Report	Selects all the data that is displayed for the run for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Compare Samples	Applicable only for runs with two or more samples. Opens the Sample Comparison dialog box. You use the options on this dialog box to compare the coverage, frequency, and read balance information for each sample that is being compared. See "Comparing Samples in a Run" on page 340.
Panel Statistics	Opens the Panel Statistics tab. The tab has options for displaying specific information based on the panel that was used for the run. See "Reviewing Panel Statistics" on page 319.
Copy	Copies all the information that is displayed for the selected run to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the run. See "Setting Read and Write Permissions" on page 326.

Table 6-1:   Runs tab, Runs pane context menu (Continued)
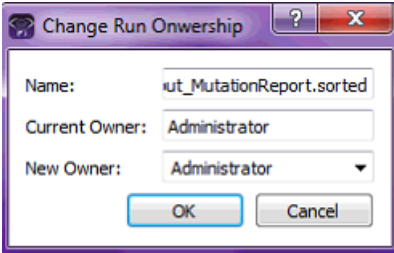
Option	Description
Change Ownership	Opens the Change Run Ownership dialog box, which displays the name of the current owner for the run, and an option (New Owner) for selecting a new owner for the run from a list of all the currently available and active users for your Geneticist Assistant instance.You can change the ownership for any run if you are an Administrator user, the current owner of the run, or if you have been granted Can Read and Can Write permissions for the run. *Figure 6-3: Change Run Ownership dialog box* **Note:** If the run was added to your Geneticist Assistant prior to V1.4, then the first time the dialog box opens, the Current Owner field is blank.
Edit Run	
Apply Variant Filter	• <Filter>: Applies the selected variant filter to all the samples in the run. When you open the Sample tab for any sample in the run, the variant filter is automatically applied. Only those sample variants that meet the filter criteria are displayed on the Sample Variant Data sub-tab of the Sample tab. The selected filter is persistent within this Geneticist Assistant instance and across instances. To clear the filter only for the current instance of Geneticist Assistant, click Clear at the bottom of the Table Filters pane. • Deactivate Filters: Permanently clears all filters that been applied for all the samples in the run for all instances of Geneticist Assistant. All variants for a selected sample are again displayed on the Sample Variant Data sub-tab.

Table 6-1:   Runs tab, Runs pane context menu (Continued)
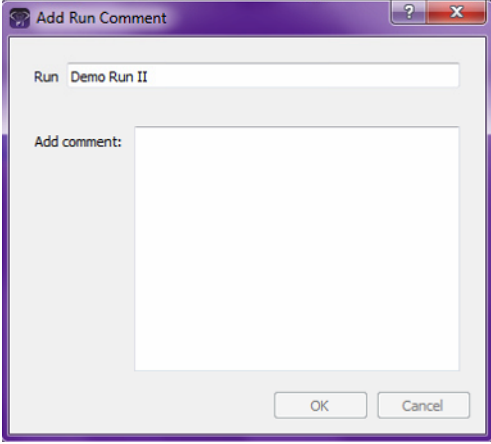
Option	Description
Add Comment	Opens the Add Run Comment dialog box. You add a comment for the selected run in the Add comment field. The comment is displayed in the Comment field on the Runs tab and the Comment field in the Comments pane on the Run tab. *Figure 6-4: Add Run Comment dialog box* **Note:** If multiple comments have been added for a run, then only the last added comment is displayed in the Comment field on the Runs tab. All the comments that have been added for a run, including the last added, are displayed in reverse chronological order in the Comments pane on the Run tab.
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 170.

Table 6-1:   Runs tab, Runs pane context menu (Continued)
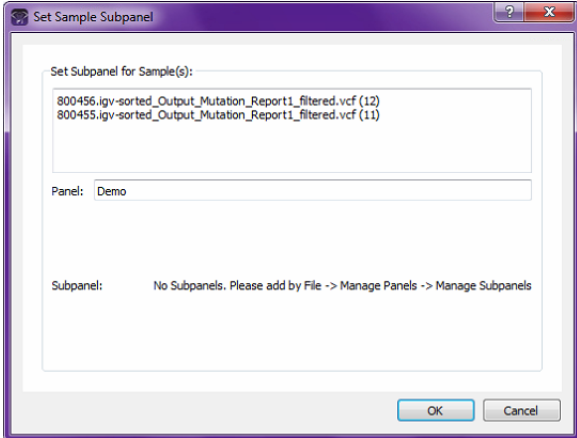
Option	Description
Set Subpanels of Selected Runs	Applicable only if you select a run for which a subpanel was not selected during the setup of the run. CTRL-click to select multiple runs. If you select multiple runs, then all the runs must use the same panel. Opens the Select a Subpanel dialog box. The dialog box displays the samples and the panel for the selected runs and a Subpanel dropdown list with all the available subpanels. *Figure 6-5: Set Sample Subpanel dialog box* Select a different subpanel on the Subpanel dropdown list, and then click OK. The subpanel is applied to all the samples in all the selected runs.
Delete Run	Delete the selected run from the Geneticist Assistant database.
Export - Contains all the sub-menu options for exporting a variant.	
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.
Save Sample Reports	Applicable for one or more runs. CTRL-click to select multiple runs. Generates an individual sample report for each sample in all the selected runs. If a single sample is selected, then a default location, report name, and report type (.csv) are provided, but you can change any or all of these values. If multiple samples are selected, then you can select only the location in which to save the report. Each report is named the same as the sample and the report format is .csv, and you cannot change either of these values. **Note:** For more information about standard reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.
Create Batch Variants Report	Opens the Batch Variants Report dialog box for generating the Batch Variants report for all the samples in a selected run. See "Managing a Batch Variants Report" on page 329.
Create Batch Coverage Report	Opens the Batch Coverage Report dialog box for generating the Batch Coverage report for all the samples in the selected run or runs. See "Generating a Batch Coverage Report" on page 335.
Sample PDF Report	
Applicable for one or more runs. CTRL-click to select multiple runs. Opens a list of Sample PDF report templates that you created and saved, or that other users created and made available to all users. Select a report template to generate an individual sample report for each sample in all selected runs. Each sample report is saved in a PDF format and is named the same as the sample for which it was generated. See "Designing a Sample PDF Report Template" on page 274 and "Generating a Sample PDF Report" on page 286.	

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

Option	Description
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Runs pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Run pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Chapter 7

Geneticist Assistant Data Review - Run Tab

Many options are available from the Run tab for reviewing your data in Geneticist Assistant. This chapter covers the following topics:

- "Overview of the Run Tab" on page 189.

- "Samples pane (Run tab)" on page 190.

- "Coverage Regions tab" on page 200.

- "The Samples Pane (Run tab) Context Menu" on page 191.

- "Status Changes pane (Run tab)" on page 204.

- "Reports pane (Run tab)" on page 207.

Overview of the Run Tab

The Run tab displays information for a selected run. 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 > Open.

- Double-click a run on the Runs tab.

Figure 7-1:   Run tab
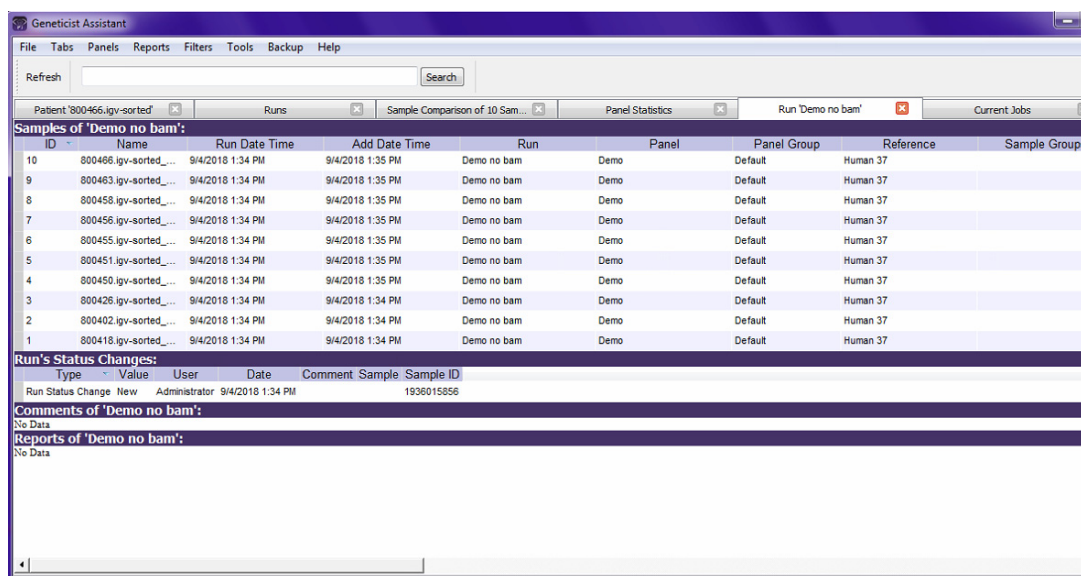
The Run tab has four panes. From top to bottom, these panes are the following:

- The Samples pane. See "Samples pane (Run tab)" on page 190.

- The Run's Status Changes pane. See "Status Changes pane (Run tab)" on page 204.

- The Comments pane. See "Comments pane (Run tab)" on page 205.

- The Reports pane. See "Reports pane (Run tab)" on page 207.

Samples pane (Run tab)

The Samples pane on the Run tab displays an entry for each sample (VCF/BAM 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 190.

- Using the context menu. See "The Samples Pane (Run tab) Context Menu" on page 191.

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. If you have not done so already, open the Run tab. (See "Overview of the Run Tab" on page 189.)

2. On the Samples pane, on the Status dropdown list, select a different sample status.

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 in the Geneticist Assistant Database" on page 55.

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 7-2:   Update Sample Status dialog box
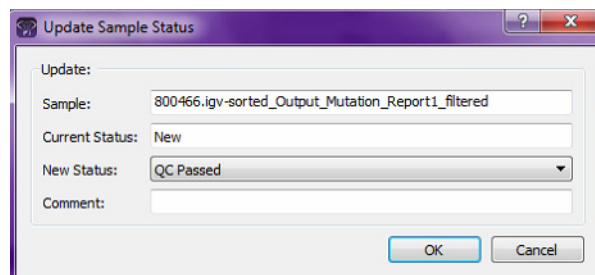
3. Optionally, in the Comment field, enter an explanation/reason for changing the sample status.

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 (Sample tab, Sample Data sub-tab)" on page 232.

4. 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.

The Samples Pane (Run tab) Context Menu

You can right-click a sample (CTRL-click to select multiple samples) in the Samples pane on the Run tab to open a context menu with the following options:

Table 7-1:   Run tab, Samples pane context menu
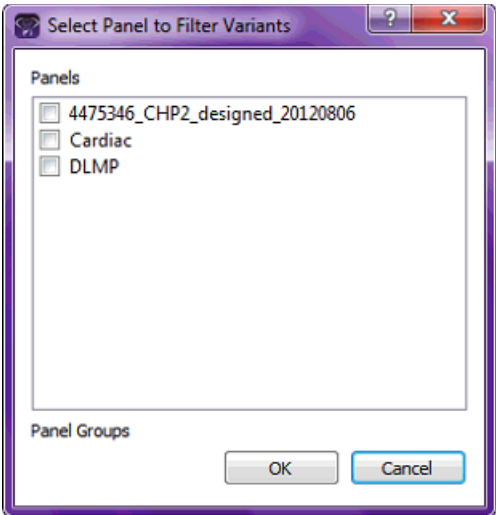
Option	Description
Open with <variant filter >	Select a filter for the sample. The Sample tab opens for the selected sample. The Sample Variant Data sub-tab is the open tab and the selected filter is applied to the tab. Only those sample variants that meet the filter criteria are displayed on the tab. The selected filter is persistent within this Geneticist Assistant instance and across instances. To clear the filter only for the current instance of Geneticist Assistant, click Clear at the bottom of the Table Filters pane. To select a different filter for the sample, or to permanently clear the for the sample, see "Apply Variant Filter" under "Edit Sample" on page 193. **Note:** For information about creating a filter template, see "To filter the data that is displayed in a tab pane" on page 32. For information about the Sample Variant Data sub-tab, see "Sample Variant Data Sub-Tab" on page 212.
Sample Details	Displayed only if a single sample is selected. Opens the Sample tab, which contains detailed information about the sample, including the variant list. See Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209.
Show Sample Variants Filtered by Panel	Displayed only if a single sample is selected. Displays all the variants in the selected sample filtered by a selected panel or panels. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel or panels in this dialog box by which to filter the variants for the selected run. *Figure 7-3: Select Panel to Filter Variants dialog box*
Coverage Regions	Displayed only if a single sample is selected. 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. See "Coverage Regions tab" on page 200.

Table 7-1:   Run tab, Samples pane context menu (Continued)

Option	Description
Show Sample Variants	Displayed only if two or more samples are selected. You can select only one of the four options. All options open the Sample Variant Data sub-tab for the Sample tab.
• All	• Displays all the variants for all selected samples.
• Distinct	• Displays all the variants that are different among the selected samples.
• All Filtered by Panel	• Displays all the variants for all selected samples filtered by the selected panel.
• Distinct Filtered by Panel	• Displays all the variants that are different among the selected samples filtered by the selected panel.
Compare Samples	Displayed only if two or more samples are selected. Opens the Sample Comparison dialog box. You use the options on this dialog box to compare the coverage, frequency, and read balance information for each sample that is being compared. See "Comparing Samples in a Run" on page 340.
Family Comparison	Displayed only if three or more samples are selected. At a minimum, you must select two parent samples and one child sample. You can always select more than one child sample. Opens the Family Comparison dialog box, in which you can specify the options to compare the variant calls in family (relation and phenotype) data. See "To compare family data" on page 197.
Panel Statistics	Opens the Panel Statistics tab. The tab has options for displaying specific information for the panel that was used for the run. See "Reviewing Panel Statistics" on page 319.
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 10, "Geneticist Assistant Reports," on page 265.
Copy	Copies all the information that is displayed for the selected sample to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the sample. See "Setting Read and Write Permissions" on page 326.
Change Ownership	Opens the Change Sample Ownership dialog box, which displays the name of the current owner for the selected sample, and an option (New Owner) for selecting a new owner for the sample. You can change the ownership for any sample if you are an Administrator user, the current owner of the sample, or if you have been granted Can Read and Can Write permissions for the sample. *Figure 7-4: Change Sample Ownership dialog box*

Table 7-1:   Run tab, Samples pane context menu (Continued)
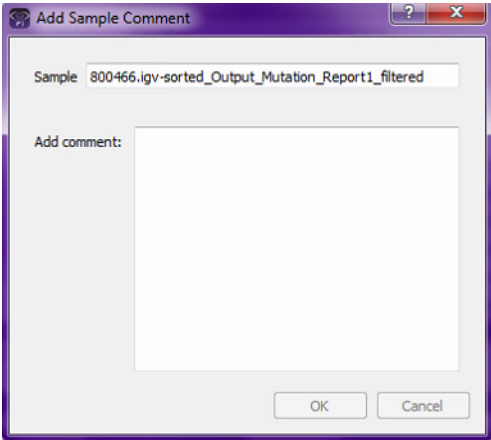
Option	Description
Edit Sample	
Apply Variant Filter	• <Filter>: Select a variant filter for the sample. The Sample tab opens for the selected sample. The Sample Variant Data sub-tab is the open tab and the selected variant filter is applied to the tab. Only those sample variants that meet the filter criteria are displayed on the tab. The selected filter is persistent within this Geneticist Assistant instance and across instances. To clear the filter only for the current instance of Geneticist Assistant, click Clear at the bottom of the Table Filters pane. • Deactivate Filters: Permanently clears all applied variant filters for the selected sample for all instances of Geneticist Assistant. All variants for the selected sample are again displayed on the Sample Variant Data sub-tab.
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 7-5: 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.

Table 7-1:   Run tab, Samples pane context menu (Continued)
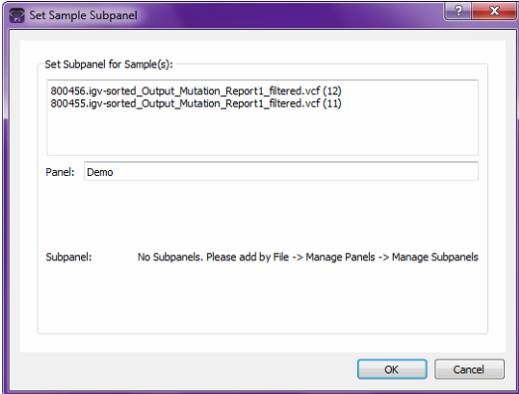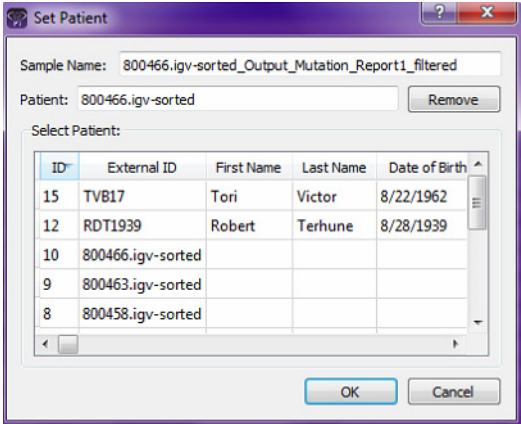
Option	Description
Set Subpanel/Set Subpanel of Selected Samples	Applicable for one or more samples for which a subpanel was not selected during the setup of the run. CTRL-click to select multiple samples. Opens the Select a Subpanel dialog box. The dialog box displays the selected sample or samples, the panel for the currently selected sample, and a Subpanel dropdown list with all the available subpanels. *Figure 7-6: Set Sample Subpanel dialog box* Select a subpanel on the Subpanel dropdown list, and then click OK. The subpanel is applied to sample or samples.
Set Patient	Opens the Set Patient dialog box. Select the patient that you are associating with the sample, and then click OK. *Figure 7-7: Set Patient dialog box* **Note:** You also have the option of associating a patient with a sample from the Sample tab. See "Patient pane (Sample Data sub-tab for the Sample tab)" on page 236.

Table 7-1:   Run tab, Samples pane context menu (Continued)
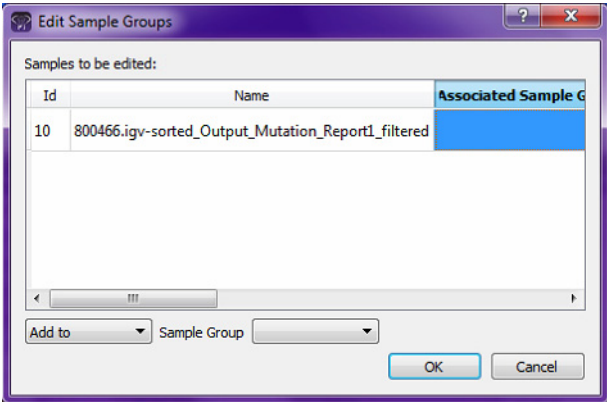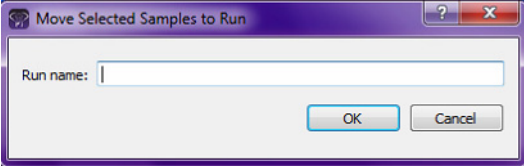
Option	Description
Edit Sample Groups	Opens the Edit Sample Group dialog box. You use the options on this dialog box to add a sample to a selected sample group, or to remove a sample from a selected sample group. **Note:** If the appropriate sample group is not available, you can add it. See "Managing Objects in the Geneticist Assistant Database" on page 55. *Figure 7-8: Edit Sample Group dialog box*
Move Samples to Run	Applicable for one or more samples. CTRL-click to select multiple samples. Opens the Move Selected Samples to Run dialog box. Enter a search string to search for the existing run in your Geneticist Assistant database that you are moving the samples to. As you enter the search string, a dynamically updated list of runs that meet your search criteria opens. You can select the appropriate run from this list. *Figure 7-9: Move Selected Samples to Run dialog box* **Note:** The search string is not case-sensitive, but the string must begin with the run name. For example, a search string of "May" or "ma" returns a run named Mayo Clinic Run II, but a search string of "clinic" does not.
Delete Sample/Delete Selected Samples	Applicable for one or more samples. CTRL-click to select multiple samples. Deletes the selected samples from the run. The samples are NOT deleted from the Geneticist Assistant database.
External - Contains all the sub-menu options for viewing the sample variants in a tool that is outside Geneticist Assistant.	
View in NextGENe	Requires you to have your NextGENe installation specified in your Geneticist Assistant settings. (See "To configure Geneticist Assistant directories" on page 59.) If selected, then Geneticist Assistant browses for the folder where the VCF file for the sample is located. If the NextGENe project file is found in the same directory as the VCF file, then the NextGENe project is opened in the NextGENe Viewer.

Table 7-1:   Run tab, Samples pane context menu (Continued)

Option	Description
View in IGV	Applicable if there is a corresponding BAM file (and not a NextGENe project file) in the same folder as the VCF file for the sample. Opens the BAM file in the third-party Integrative Genomics Viewer (IGV).
Export - Contains all the sub-menu options for exporting a variant.	
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.
Save Sample Reports	Applicable for one or more samples. CTRL-click to select multiple samples. Generates an individual sample report for each selected sample. If a single sample is selected, then a default location, report name, and report type (.csv) are provided, but you can change any or all of these values. If multiple samples are selected, then you can select only the location in which to save the report. Each report is named the same as the sample and the report format is .csv, and you cannot change either of these values. **Note:** For more information about standard reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.
Create Batch Variants Report	Opens the Batch Variants Report dialog box for generating the Batch Variants report for the selected sample or samples. (CTRL-click to select multiple samples.) See "Managing a Batch Variants Report" on page 329.
Create Batch Coverage Report	Opens the Batch Coverage Report dialog for generating the Batch Coverage report the selected sample or samples. (CTRL-click to select multiple samples.). See "Generating a Batch Coverage Report" on page 335.
Reports	
Add All to Custom Report	Adds only the data that you select to the custom report. You must CTRL-click to select multiple items for inclusion in the report. See "Generating a Custom Report" on page 271.
Add Selected Samples to Custom Report	Adds all the data that is currently displayed in the pane to the custom report. See "Generating a Custom Report" on page 271.
Sample PDF Report	
Applicable for one or more samples. CTRL-click to select multiple samples. Opens a list of custom report templates that you created and saved, or that other users created and made available to all users. Select a report template to generate an individual sample report for each selected sample. Each sample report is saved in a PDF format and is named the same as the sample for which it was generated. See "Designing a Sample PDF Report Template" on page 274 and "Generating a Sample PDF Report" on page 286.	
Load Filter	
Manually	Opens the Filters table for the Samples pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Samples pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Table 7-1: Run tab, Samples pane context menu (Continued)

Option	Description
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39.
	Note: If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
\<Saved Pane Configuration Name\> - Saved by \<User Name\>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.

To compare family data

If the samples for a run are from a family, then you can use the Family Comparison option to compare the samples based on the relationships and phenotypes for each sample. At a minimum, you must select two parent samples and one child sample. You can always select more than one child sample.

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

2. In the Samples pane, CTRL-click to select the family data that you are comparing.

At a minimum, you must select two parent samples and one child sample. You can always select more than one child sample.

3. Right-click any selected sample, and on the context menu that opens, click Family Comparison.

The Family Comparison dialog box opens. If a patient is associated with a selected sample, then patient Relationship (Father, Mother, or so on) is displayed on the Relationship dropdown list; otherwise, you must select the appropriate relationship for the patient. Likewise, if the phenotype was included with the patient information, then the phenotype is automatically displayed on the Phenotype dropdown list for the sample, where Affected indicates that the patient has the phenotype and Unaffected indicates that the patient does not have the phenotype. If the phenotype is not automatically displayed for a sample, then you must select the appropriate value. See Figure 7-10 on page 198.

Figure 7-10:   Family Comparison dialog box
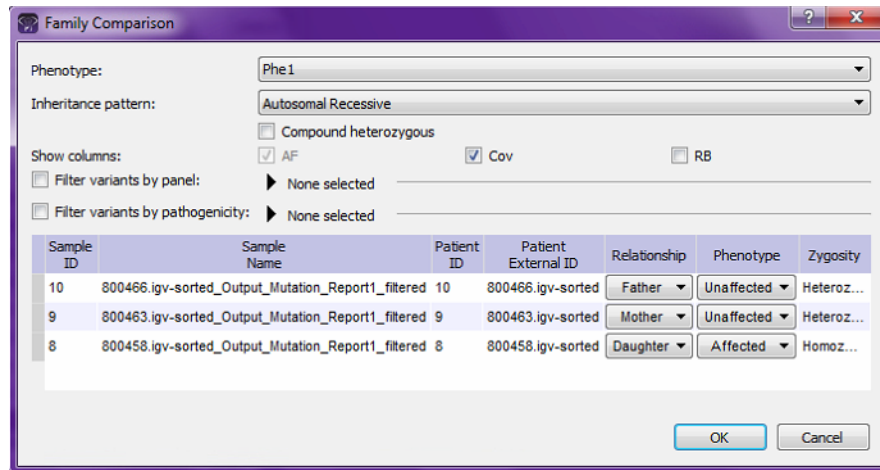
4. Do any or all of the following:

    •   On the Phenotype dropdown list at the top of the dialog box, select a different
        phenotype.

    •   On the Inheritance pattern dropdown list, select the appropriate value for the family,
        and if applicable, based on the selected Inheritance pattern, select Compound
        heterozygous.

    > *Select Compound heterozygous to carry out compound heterozygous filtering. The
    > filtering results are displayed in the Family Data Comparison Results pane. the
    > results show all possible combinations of two heterozygous variants in a gene if
    > the variants meet the relationship and phenotype settings for the parent samples.
    > For example, if both the Mother and Father are Unaffected but a Son is Affected,
    > then one heterozygous variant in the affected son must come from each parent.*

    > *After you select an Inheritance pattern, the zygosity for each sample is
    > automatically adjusted based on the selected inheritance pattern and the
    > relationship and phenotype for the sample. If you select an incorrect Inheritance
    > pattern based on the relationships and phenotypes, then "Error" is displayed
    > instead of a valid zygosity value.*

    •   Indicate which columns to show or hide in addition to the default columns that are
        displayed for the family data comparison results: AF (Allele Frequency), Cov
        (Coverage), and/or RB (Read Balance).

    •   To display all the variants in each sample filtered by a selected panel, select Filter
        variants by panel, and then on the Panels dropdown list, select the appropriate panel.

    •   To display all the variants in each sample filtered by a selected pathogenicity, select
        Filter variants by pathogenicity, and then on the Pathogenicities dropdown list, select
        the appropriate pathogenicity.

5. Click OK.

The family data comparison is carried out. The Family Comparison tab opens. The left side of the Family Comparison Results pane shows sample-specific information. The results are shown using standard pedigree symbols. If the patient that is associated with a sample is affected, then the pedigree symbol is shaded; otherwise, the symbol is not shaded. You can hover your cursor over a column header to view specific details for the selected sample. The right side of the tab shows variant detail information. See Figure 7-11 and Figure 7-12 below.

Comparison of family data can take 30 minutes or more depending on a variety of factors, including the number of the samples that are being compared and the number of variants for each sample.

Figure 7-11:   *Viewing information for a family comparison result*
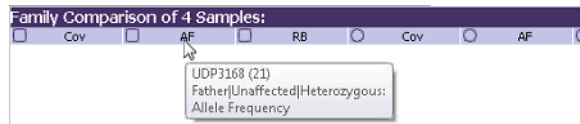
Figure 7-12:   *Family Comparison Results pane*

Coverage Regions tab

The Coverage Regions 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.
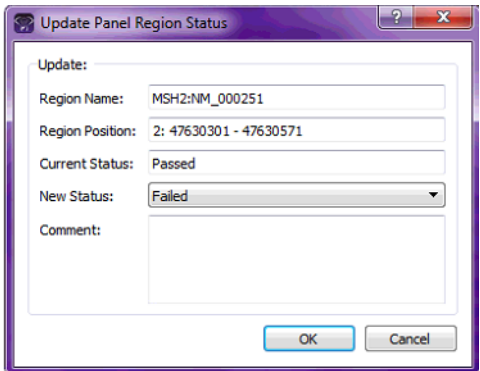
Figure 7-13:   Coverage Regions tab

Table 7-2:   Coverage Regions tab

Column	Description
ID	System-generated ID for the region.
Region Name	The region description from Column 4 of the BED file that was used to create the panel.
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 "To specify the global Quality Control settings" on page 64.
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 "To specify the global Quality Control settings" on page 64..

Table 7-2: Coverage Regions tab (Continued)

Column	Description
Minimum Coverage Threshold	The minimum coverage (number of reads aligned) that is 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 "To specify the global Quality Control settings" on page 64.
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 7-14: 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 in the Geneticist Assistant Database" on page 55.
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.
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.
Exon Number	The coding exon number for the region.
SD % Covered	The standard deviation for the "% Covered" value across all samples for the panel.
SD Average Coverage	The standard deviation for the "Average Coverage" value across all samples for the panel.

Table 7-2:   Coverage Regions tab (Continued)

Column	Description
SD Minimum Coverage	The standard deviation for the "Minimum Coverage" value across all samples for the panel.

The context menu for the Coverage Regions 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 212.) You can also right-click anywhere on the Coverage Regions tab to open a context menu with the following options:

Table 7-3:   Coverage Regions tab context menu

Option	Description
Add Selected Regions to Action List	Adds the selected panel region to an Action List. CTRL-click to select multiple regions. See "Creating an Action List" on page 338.
Add Selected Regions to Custom Report	Selects all the data that is displayed for the selected panel region for inclusion in a custom report. CTRL-click to select multiple regions. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Copy	Copies all the coverage information that is displayed for the selected run to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.
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 10, "Geneticist Assistant Reports," on page 265.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39.
	Note: If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Coverage Regions pane. See "To filter the data that is displayed in a tab pane" on page 32.

Table 7-3:   Coverage Regions tab context menu (Continued)

Option	Description
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Coverage Regions pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Status Changes pane (Run tab)

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 first run. You can right-click anywhere on this 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 the statuses for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39.
	Note: If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Run Status Changes pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Run Status Changes pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Comments pane (Run tab)

The Comments pane on the Run tab shows all the comments that have been added for the run in reverse chronological order. If one or more comments are displayed in the Comments pane, then you can right-click anywhere on this pane to open a context menu with the following options:

The context menu for the Comments pane is not enabled if the pane is blank. To add a comment for the run and enable this context menu, see "The Runs Tab Context Menu" on page 181.

Option	Description
Add Run Comment	Opens the Add Comment dialog box. You add a comment for the selected run in the Add comment field. The comment is displayed in the Comment field on the Runs tab and the Comment field in the Comments pane on the Run tab. *Figure 7-15: Add Run Comment dialog box* **Note:** If multiple comments have been added for a run, then only the last added comment is displayed in the Comment field on the Runs tab. All the comments that have been added for a run, including the last added, are displayed in reverse chronological order in the Comments pane on the Run tab.

Option	Description
Delete Run Comment	Opens the Delete Comment dialog box. The dialog box displays the name of the selected run and the comment that is being deleted. Click OK to delete the selected comment. *Figure 7-16: Delete Comment dialog box*
Add All to Custom Report	Selects all the information that is displayed for all the statuses for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Run Status Changes pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Run Status Changes pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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 includes 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 154.

For detailed information about generating and saving both standard and custom Run reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.

Chapter 8

Geneticist Assistant Data Review - Sample Tab

Many options are available from the Sample tab for reviewing your data in Geneticist Assistant.

This chapter covers the following topics:

Overview of the Sample Tab

The Sample tab displays information for a selected sample. To open the Sample tab, do one of the following:

- Right-click a run on the Runs tab, and on the context menu that opens, click Run > Open all Samples with, and then select a filter template. After the Sample tab is populated based on the selected filter template, you can clear the template to display all sample variant data for the run.

- Right-click a sample in the Samples pane on the Run tab, and on the context menu that opens, select Sample Details.

- Double-click a sample in the Samples pane on the Run tab.

- Right-click a sample on the Samples sub-tab on the Patient tab, and on the context menu that opens, select Sample Details.

- Double-click a sample on the Samples sub-tab on the Patient tab.

- Right-click a sample in the Associated Samples pane on the Variant tab, and on the context menu that opens, select Sample Details.

- Double-click a sample in the Associated Samples pane on the Variant tab.

Figure 8-1: Sample tab, Sample Variant Data sub-tab

The Sample tab has four sub-tabs:

- The Sample Variant Data sub-tab. See "Sample Variant Data Sub-Tab" on page 212.

- The Sample Data sub-tab. See "Sample Data Sub-Tab" on page 232.

- The Compound Variant Interpretation sub-tab. See "Compound Variant Interpretation Sub-Tab" on page 240.

- The Submission sub-tab. See "Submission Sub-Tab" on page 241.

Sample Variant Data Sub-Tab

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

Figure 8-2:   Sample tab, Sample Variant Data sub-tab

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. By default, two types of information are always displayed on the sub-tab: variant annotation information and global variant count and frequency information. The pane also displays statistical information if a sample group was used for the run and statistics were calculated for the sample group. A variety of options are available for working with a variant that is displayed on this sub-tab, and a context menu is also available. See:

- "Variant annotation information" on page 213.

- "Global variant count and frequency information" on page 218.

- "Sample group statistical information" on page 219.

- "Variant options on the Sample Variant Data sub-tab" on page 220.

- "Sample Variant Data sub-tab context menu" on page 222.

The information that is displayed on the Sample Variant Data sub-tab was divided into three separate 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 which columns to hide in a pane. See "To show/hide columns in a tab pane" on page 39.*
- *Remember, you can specify how the information about the variants is displayed on the sub-tab. See "To specify variant preferences" on page 70.*
- *Selected columns from the various reference information panes on the Variant tab can also be displayed here. See Chapter 9, "Geneticist Assistant Data Review - Variant Tab," on page 243.*

Variant annotation information

The following table lists the variant annotation information that is displayed on the Sample Variant Data sub-tab.

Many of the options that are listed in the variant annotation information columns are also available on the Sample Variant Data sub-tab context menu.

Table 8-1: Sample Variant Data 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.

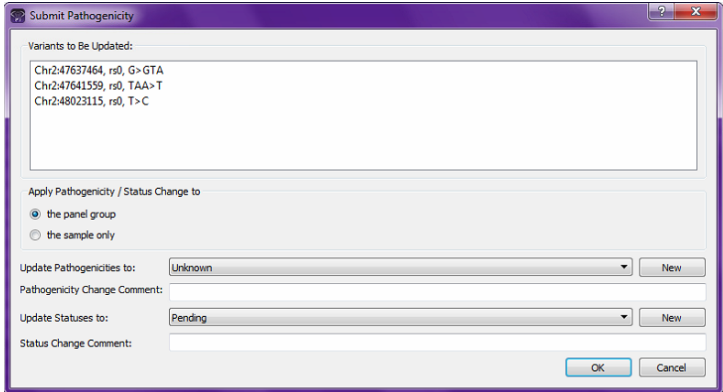*Table 8-1:   Sample Variant Data sub-tab: Variant Annotation information (Continued)*

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, then you can set the status to Unknown.) When you update the pathogenicity and/or status, you can apply the changes to: • the panel group - Applies the changes to the variant for the indicated panel group.The next time that the variant is detected in another sample that has been assigned to this panel group, then this is the pathogenicity that is displayed for the variant. • the sample only - Applies the changes to the variant only for the currently selected sample. You can also update the pathogenicity status and/or add comments for the changes. By default, the status is always set to an initial value of Pending. *Figure 8-3: Submit Pathogenicity dialog box* **Note:** You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box, or use the context menu option. See "Sample Variant Data sub-tab context menu" on page 222. The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five pathogenicities—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities, either by clicking New to open the Add New Pathogenicity dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. • By default, only two statuses—Pending and Confirmed—are available. You have the option of adding custom statuses, either by clicking New to open the Add New Pathogenicity Status dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. • Optionally, you can add supporting information for this pathogenicity change. See "To add supporting information for a variant" on page 256. • You always have the option of updating just the pathogenicity status for a variant. See "Pathogenicity Status" on page 215. • You can also update the pathogenicity on the Patient tab, Variants sub-tab. See "Patient tab, Variants sub-tab" on page 158.

Table 8-1:   Sample Variant Data sub-tab: Variant Annotation information (Continued)
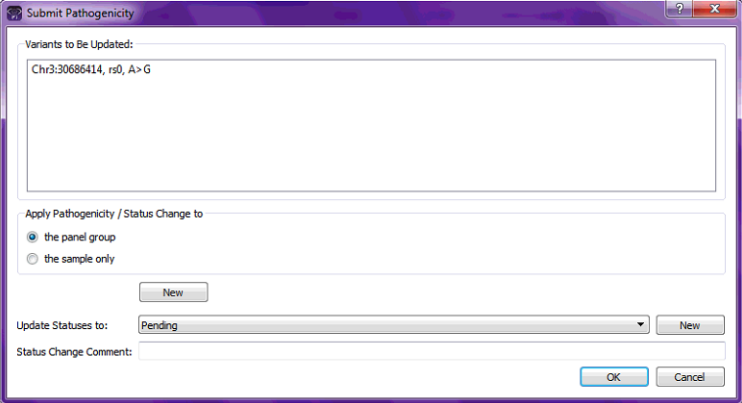
Column	Description
Pathogenicity Date Time	The date and time that the currently displayed pathogenicity value was assigned to the variant.
Pathogenicity Status	The status of the selected pathogenicity. You must manually set this value. By default, only two statuses—Pending and Confirmed—are available, and the pathogenicity status is always set to an initial value of Pending. You must review the information for the variant and then decide what the pathogenicity status should be. To open the Submit Pathogenicity dialog box, double-click in this column for a selected variant.You can update the pathogenicity status for the selected variant in this dialog box. When you update the pathogenicity status, you can apply the changes to:
	• the panel group - Applies the changes to the variant for the indicated panel group.The next time that the variant is detected in another sample that has been assigned to this panel group, then this is the pathogenicity status that is displayed for the variant.
	• the sample only - Applies the changes to the variant only for the currently selected sample.
	You can also add a comment for the status change.
	You have the option of adding custom statuses, either by clicking New to open the Add New Pathogenicity Status dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55.
	Figure 8-4: 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 Status column to open the Update Variant Pathogenicity Status dialog box or use the option that is available on the context menu for the sub-tab. See "Sample Variant Data sub-tab context menu" on page 222.
	• You can also update the Pathogenicity Status for a variant on the Patient tab, Variants sub-tab. See "Patient tab, Variants sub-tab" on page 158.
Pathogenicity Status Date Time	The date and time that the currently displayed pathogenicity status was assigned to the variant.
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 70.

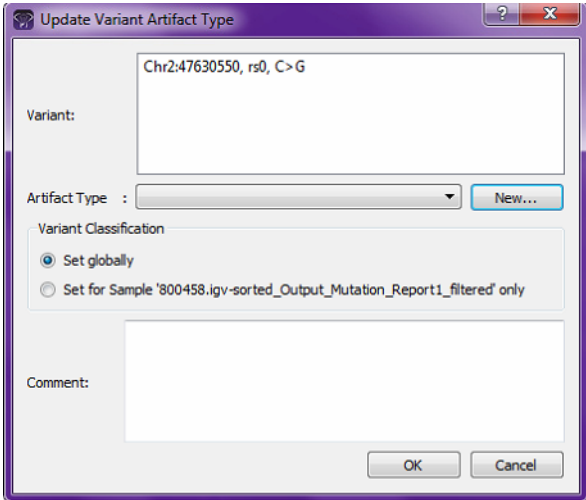*Table 8-1:   Sample Variant Data sub-tab: Variant Annotation information (Continued)*
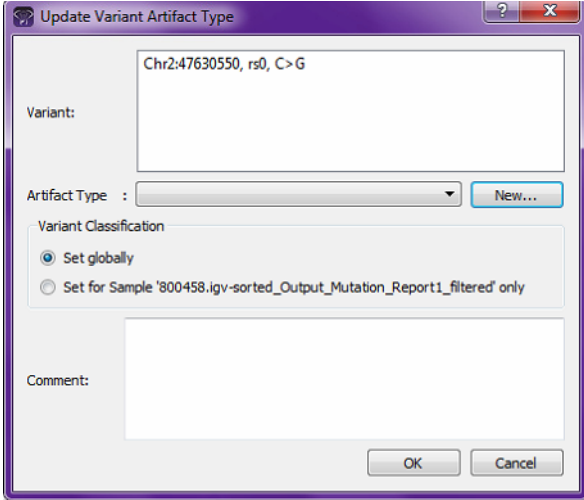
Column	Description
Artifact Type	The artifact type for the selected variant. You must manually set this value. Double-click in this column for the selected variant to open the Update Artifact Type dialog box. You specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type. *Figure 8-5: 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. **Note:** The default value for Variant Classification is determined by the setting on the Variant Preferences tab on the Settings dialog box. See "To specify variant preferences" on page 70. The following information is relevant to updating the artifact type for a variant: • 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 from the Runs tab or the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • You can also double-click in the Artifact Type column to open the Update Variant Artifact Type dialog box, or use the context menu option. See "Sample Variant Data sub-tab context menu" on page 222. • You have the option of adding custom artifact types, either by clicking New to open the Add New Artifact dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55.
Patient 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.
Gene	The gene on which the variant is located.
Gene Strand	The strand (+ or -) for the gene in which the variant is located.
Read Balance	The balance in forward and reverse oriented reads with the variant as reported in the VCF file.

Table 8-1:   Sample Variant Data sub-tab: Variant Annotation information (Continued)
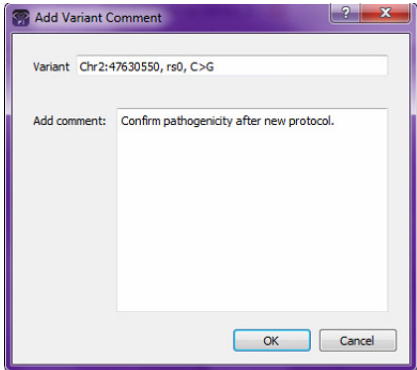
Column	Description
Exon Number	The exon within the gene in which the variant is located. Includes non-coding exons. "Intron" is displayed for intronic variants.
Transcript	The accession number for the preferred mRNA transcript that is used for the HGVS nomenclature columns.
Protein	The accession number for the protein transcript that is associated with the preferred mRNA transcript and is used for the HGVS nomenclature columns.
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. (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 single-click twice in this field, and then add or edit a comment as needed. After you click out of the field, the Add Variant Comment dialog box opens. The comment is displayed in the Variant Comment field in the Variants pane on the Search Results 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 8-6: 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 here and on the Variants sub-tab on the Patient tab, the Sample Variant Data sub-tab in 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. You can also add comments for a variant with an option on the context menu for this sub-tab. See "Add Variant Comment" on page 224.
Comment Date Time	The date and time that the currently displayed comment (The last added comment for the variant) was added for the variant.

Table 8-1:   Sample Variant Data sub-tab: Variant Annotation information (Continued)

Column	Description
Panel	The panel that was used for the run.
Panel Group	The panel group to which the panel is assigned.
Trans	Indicates whether the variant is Transition (Ti) or Transversion (Tv).
Sample	The sample name.
CDS number	The coding exon number.
Genotype	The variant genotype.
Report	Displays a checkbox. For each variant that is to be included in a report, click in the checkbox to select the variant. **Tip:** You can define variant filtering options to filter for variants only where Report = true or Report = false.
Interpretation	If an interpretation has been defined for this single variant, then the full interpretation is displayed in this column. If more than one interpretation has been defined for the variant, then the last defined interpretation is displayed.
Coverage Region Status	Indicates whether the region passed or failed the Quality Control (coverage) settings that were specified for the run's panel.

Global variant count and frequency information

The following table lists the global variant count and frequency information that is displayed on the Sample Variant Data sub-tab.

Table 8-2:   Sample Variant Data sub-tab: Global Variant Count and Frequency information

Column	Description
Samples Per Panel	The total number of samples submitted per 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 Run	The number of times that the variant was detected across all samples in the run.
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 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.

Table 8-2:   Sample Variant Data sub-tab: Global Variant Count and Frequency information (Continued)

Column	Description
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.
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 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.

Sample group statistical information

The following table lists the sample group statistical information that is displayed on the Sample Variant Data sub-tab.

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 "Sample Variant Data sub-tab context menu" on page 222.

Table 8-3:   Sample Group Statistics Columns on the Sample Variant Data sub-tab

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 Mean	The mean 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.

Table 8-3: Sample Group Statistics Columns on the Sample Variant Data sub-tab (Continued)

Column	Description
Variant Frequency Min	The minimum frequency at which the variant was detected in the sample group. Any value $\leq$ 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 $\leq$ 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 $\leq$ 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 $\leq$ 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.

Variant options on the Sample Variant Data sub-tab

You can do the following for a variant that is displayed on the Sample Variant Data sub-tab on the Sample 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 70.

- If the variant is located where there are overlapping genes, then you can select a different gene for the variant on the Gene dropdown list. If you select a different gene, then the gene that is displayed on the tab for the current Geneticist Assistant session is temporarily changed. After you close and then re-open Geneticist Assistant, the selected gene is reset to the preferred gene. To permanently change the gene that is used for variant, you can add a preferred gene for the panel. See "To set or delete a preferred gene for a panel" on page 112.

- You can select a different transcript for the variant on the Transcript dropdown list. If you select a different transcript, then the transcript that is displayed on the tab for the current Geneticist Assistant session is temporarily changed. After you close and then re-open Geneticist Assistant, the selected transcript is reset to the preferred transcript.

> *To permanently change the transcript that is used for a gene or genes, you can do one of two things: You can right-click the variant, and on the context menu that opens, click Edit Variant > Update Preferred Transcript (see "Sample Variant Data sub-tab context menu" on page 222), or you can add a preferred transcript list for the panel. See "To set or delete a preferred transcript list for a panel" on page 110.*

- 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 Add Variant Comment dialog box opens.

Figure 8-7:   *Add Variant Comment dialog box*
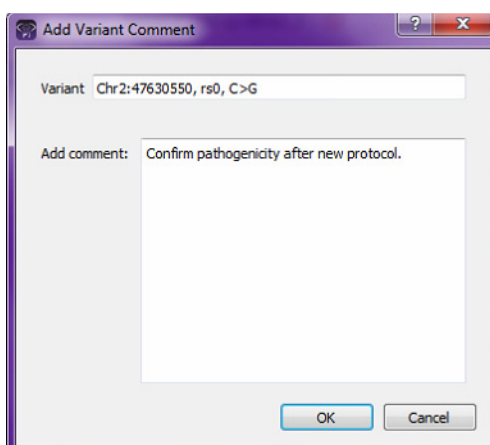
The dialog box displays the variant for which you are adding or editing the comment and the current comment. You can edit the current comment, or leave it as-is, and then click OK to close the dialog box and save the variant comment.

> *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 224.*

- You can single click twice in the Type field, the Zygosity field, the HGVS Genomic field, the HGVS Coding field, and/or the HGVS Protein field to select the current value in the field and edit it. If the Type field is blank, then you can enter a 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 251. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in Geneticist Assistant.*

- You can single click twice in the Interpretation field to select the current interpretation in the field and edit it, or if the field is blank, enter a new interpretation. After you add or edit an interpretation and then click out of the field, the Add Variant Interpretation dialog

box opens.

Figure 8-8:   Add Variant Comment dialog box
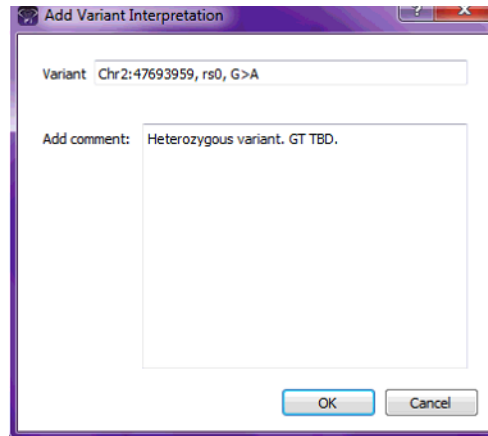
The dialog box displays the variant for which you are adding or editing the interpretation
and the current interpretation. You can edit the current interpretation, or leave it as-is,
and then click OK to close the dialog box and save the variant interpretation.

Sample Variant Data sub-tab context menu

You can right-click any column header on the Sample Variant Data sub-tab to open a context
menu that, in addition to the standard column header context menu options, contains a
Manage Sample Group Statistics Columns option. This option opens the Select Sample
Groups dialog box.

Figure 8-9:   Select Sample Groups dialog box
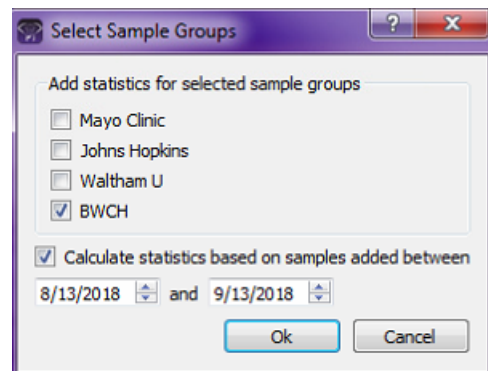
- If you used sample groups for a run, then you can indicate the date range for which to
  calculate statistics based on the added samples.

- If you did not use specific sample groups for a run, then do not select these sample groups,
  or if applicable, clear the selections for these groups, which hides the corresponding
  Sample Group Statistics columns from the sub-tab display.

You can also right-click a variant in the Variants pane on the sub-tab to open a context menu with the following options:

Table 8-4:   Sample tab, Sample Variant Data sub-tab context menu
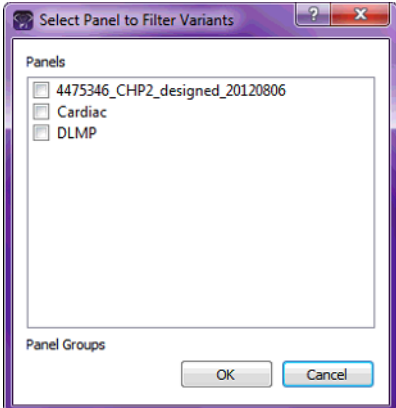
Option	Description
Variant Details	Opens the Variant tab, which displays detailed information about the variant. See Chapter 9, "Geneticist Assistant Data Review - Variant Tab," on page 243.
Show Variants Filtered by Panel	Displays all the variants in the selected sample filtered by a selected panel or panels. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel or panels in this dialog box by which to filter the variants for the selected run. *Figure 8-10: Select Panel to Filter Variants dialog box*
Show Samples with Selected Variants	Applicable for one or more variants. CTRL-click to select multiple variants. Two options available: • All variants - Opens the Samples with all variants pane, which displays a list of all the samples in your Geneticist Assistant database that contain all the selected variants. You can right-click a sample in the list to open a context menu with sample-specific options. See "The Samples Pane (Run tab) Context Menu" on page 191. • Any variant - Opens the Samples with any variants pane, which displays a list of all the samples in your Geneticist Assistant database that contain any of the selected variants. You can right-click a sample in the list to open a context menu with sample-specific options. See "The Samples Pane (Run tab) Context Menu" on page 191.

Table 8-4:   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. You can also CTRL-click to select multiple variants and add the same comment to all the selected variants in a single step. The comment is displayed in the Variant Comment field in the Variants pane on the Search Results 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 8-11: 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.
\<Add Variant Interpretation\>	Select a variant, or CTRL-click to select multiple variants. • If a single variant is selected, then the option is Add Single Variant Interpretation, and the Add Single Variant Interpretation dialog box opens. • If two or more variables are selected, then the option is Add Compound Variant Interpretation and the Add Compound Variant Interpretation dialog box opens. See "Adding a Variant Interpretation" on page 342.

Table 8-4:   Sample tab, Sample Variant Data sub-tab context menu (Continued)

Option	Description
Update 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, then you can set the status to Unknown.) When you update the pathogenicity and/or status, you can apply the changes to: • the panel group - Applies the changes to the variant for the indicated panel group.The next time that the variant is detected in another sample that has been assigned to this panel group, then this is the pathogenicity that is displayed for the variant. • the sample only - Applies the changes to the variant only for the currently selected sample. You can also update the pathogenicity status and/or add comments for the changes. By default, the status is always set to an initial value of Pending. *Figure 8-12: Submit Pathogenicity dialog box* **Note:** You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box. The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant: • By default, only five pathogenicities—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities, either by clicking New to open the Add New Pathogenicity dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. • By default, only two statuses—Pending and Confirmed—are available. You have the option of adding custom statuses, either by clicking New to open the Add New Pathogenicity Status dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. • Optionally, you can add supporting information for this pathogenicity change. See "To add supporting information for a variant" on page 256. • You always have the option of updating just the pathogenicity status for a variant. See "Pathogenicity Status" on page 215. • You can also update the pathogenicity on the Patient tab, Variants sub-tab. See "Patient tab, Variants sub-tab" on page 158.

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

Option	Description
Update Pathogenicity Status	Select a variant, or CTRL-click to select multiple variants and then select this option to open the Submit Variant Pathogenicity Status dialog box. You can update the pathogenicity status for the selected variants in this dialog box. When you update the pathogenicity status, you can apply the changes to: • the panel group - Applies the changes to the variant for the indicated panel group. The next time that the variant is detected in another sample that has been assigned to this panel group, then this is the pathogenicity status that is displayed for the variant. • the sample only - Applies the changes to the variant only for the currently selected sample. You can also add a comment for the status change. You have the option of adding custom statuses, either by clicking New to open the Add New Pathogenicity Status dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55. *Figure 8-13: Submit 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 also update the Pathogenicity Status for a variant on the Patient tab, Variants sub-tab. See "Patient tab, Variants sub-tab" on page 158.

Table 8-4:   Sample tab, Sample Variant Data sub-tab context menu (Continued)

Option	Description
Update 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. *Figure 8-14: 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. **Note:** The default value for Variant Classification is determined by the setting on the Variant Preferences tab on the Settings dialog box. See "To specify variant preferences" on page 70. The following information is relevant to updating the artifact type for a variant: • 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 from the Runs tab or the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • You have the option of adding custom artifact types, either by clicking New to open the Add New Artifact dialog box and entering a new value, or from the Manage Objects function. See "Managing Objects in the Geneticist Assistant Database" on page 55.

Table 8-4:   Sample tab, Sample Variant Data sub-tab context menu (Continued)
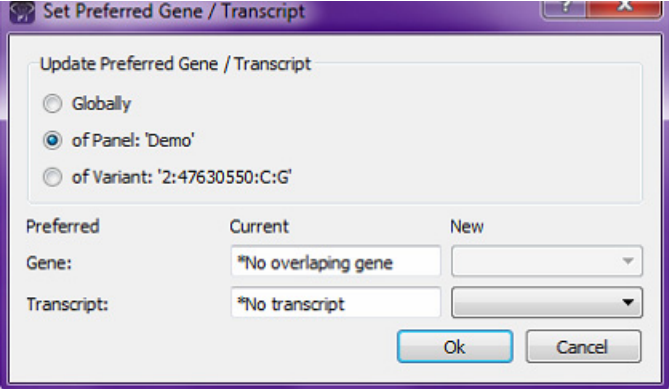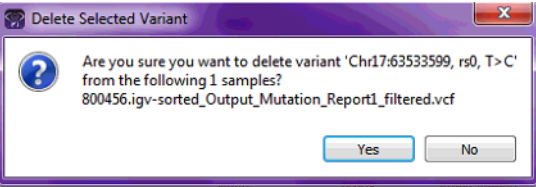
Option	Description
Update Preferred Transcript	Opens the Set Preferred Gene/Transcript dialog box. You can select a different preferred gene and/or transcript for the variant. When you select a different gene/ preferred transcript, you can set the preferences for one of the following: • globally - for the variant in all panels and panel groups. • of panel - for the variant only when in the indicated panel. • of Variant - only for this particular variant and not for any other positions within this panel or panel group. *Figure 8-15: Set Preferred Gene/Transcript dialog box* **Note:** When you select a different transcript here for a panel, then the preferred transcript that is used for a gene or genes is permanently changed for the panel. You can also permanently change the preferred transcript by adding a preferred transcript list for the panel. See "To set or delete a preferred transcript list for a panel" on page 110. To temporarily change the preferred transcript for a gene or genes, you simply select the transcript on the Transcript dropdown list on a tab or sub-tab.
Delete Variant from This Sample	Deletes the selected variant from this sample only. *Figure 8-16: Delete Selected Variant dialog box*

Table 8-4:   Sample tab, Sample Variant Data sub-tab context menu (Continued)
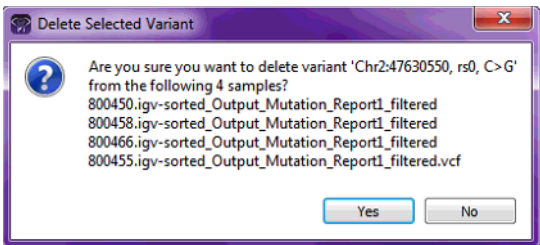
Option	Description
Delete Variant from All Samples	Deletes the selected variant from all samples in which it was found. *Figure 8-17: Delete Selected Variant dialog box* ![Delete Selected Variant dialog box: "Are you sure you want to delete variant 'Chr2:47630550, rs0, C>G' from the following 4 samples? 800450.igv-sorted_Output_Mutation_Report1_filtered 800458.igv-sorted_Output_Mutation_Report1_filtered 800466.igv-sorted_Output_Mutation_Report1_filtered 800455.igv-sorted_Output_Mutation_Report1_filtered.vcf" with Yes and No buttons]
External - Contains all the sub-menu options for viewing the sample variants in a tool that is outside Geneticist Assistant.	
View in NextGENe	Requires you to have your NextGENe installation specified in your Geneticist Assistant settings. (See "To configure Geneticist Assistant directories" on page 59.) If selected, then Geneticist Assistant browses for the folder where the VCF file for the sample is located. If the NextGENe project file is found in the same directory as the VCF file, then the NextGENe project is opened in the NextGENe Viewer, with the focus set to the position for the selected variant.
View in IGV	Applicable if there is a corresponding BAM file (and not a NextGENe project file) in the same folder as the VCF file for the sample. Opens the BAM file in the third-party Integrative Genomics Viewer (IGV) with the focus set to the selected variant.
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 third-party (Alamut, IGV, OMIM) settings" on page 74.
View in UCSC	Opens the variant for viewing in the UCSC Genome Browser.

Table 8-4:   Sample tab, Sample Variant Data sub-tab context menu (Continued)
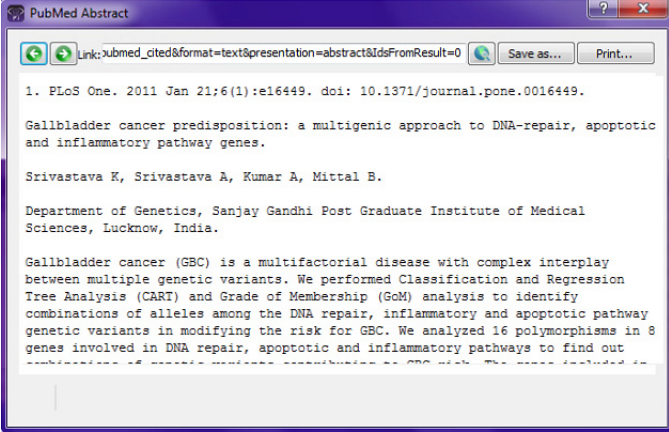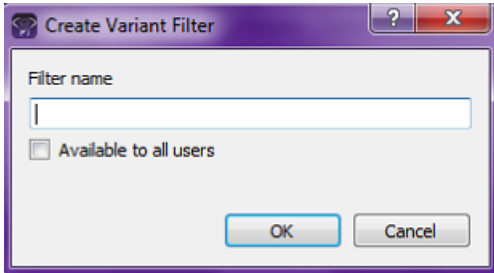
Option	Description
View PubMed Abstract	Searches PubMed for the variant and displays the appropriate abstract in the PubMed window. *Figure 8-18: PubMed window* You can do the following for the abstract: • In the Link field, enter a different PubMed URL, and then click the Go To icon to navigate to abstract. • 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. CTRL-click 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. CTRL-click to select multiple variants. See "Creating an Action List" on page 338.
Add Selected Variants to Custom Report	Selects all the data that is displayed for the selected variants for inclusion in a custom report. CTRL-click to select multiple variants. See Chapter 10, "Geneticist Assistant Reports," on page 265.
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 10, "Geneticist Assistant Reports," on page 265.
Copy	Copies all the information that is displayed for the selected variant to your client's clipboard. You can use standard Paste commands to paste this copied information into a third-party application such as Notepad.

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

Option	Description
Create Variant Filters from Selected Variants	Create a filter based on the selected variants. You can select a single variant, or CTRL-click to select multiple variants to open the Create Variant Filter dialog box. Name the filter and indicate if the filter is to be available to all users. You, and if applicable, other users can use the Load Filter Template function to apply this variant filter to a selected sample or samples. (See "To load a saved filter template" on page 38.)

Figure 8-19: Create Variant Filter dialog box

When the Sample tab opens for a sample to which the filter has been applied, then the Sample Variant Data sub-tab displays only those variants that meet the filter criteria.

Load Filters

Option	Description
Manually	Opens the Filters table for the Variants pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Variants pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Load Columns

Option	Description
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.

Sample Data Sub-Tab

The Sample Data sub-tab contains six panes—Status Changes, Comments, Patient, Related Patients, Variant Interpretations, and Reports—that provide different information about the selected sample.

Figure 8-20:   Sample tab, Sample Data sub-tab
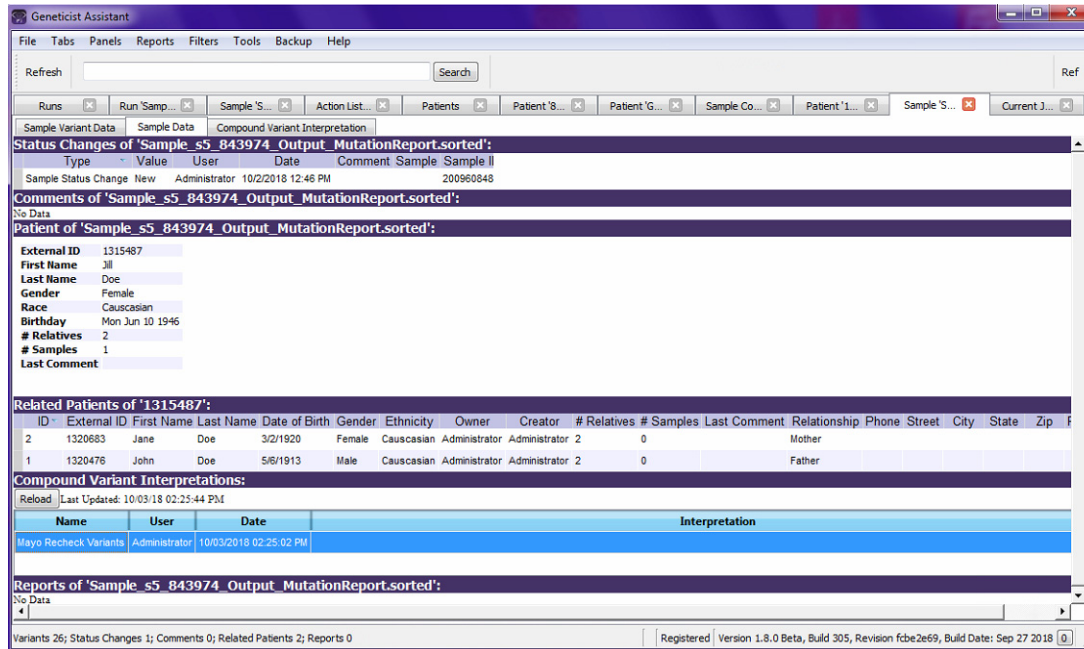
With the exception of the Compound Variant Interpretations pane, a context menu is available for each pane. See:

- "Status Changes pane (Sample tab, Sample Data sub-tab)" below.

- "Comments pane (Sample Data sub-tab on the Sample tab)" on page 234.

- "Patient pane (Sample Data sub-tab for the Sample tab)" on page 236.

- "Related Patients pane (Sample Data sub-tab for the Sample tab)" on page 237.

- "Compound Variant Interpretations pane" on page 238.

- "Reports pane (Sample data sub-tab on the Sample tab)" on page 238.

Status Changes pane (Sample tab, Sample Data sub-tab)

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 sample status set to New.

Remember, you can change the status for a sample on the Run tab. See "To set the sample status" on page 190.

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

Table 8-5:   Sample tab, Sample Data sub-tab, Status Changes 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 10, "Geneticist Assistant Reports," on page 265.
Copy	Copies all the information that is displayed for the selected sample to your client's clipboard. You can use standard Paste commands to paste this copied information into a third-party application such as Notepad.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Status Changes pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Status Changes pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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. If one or more comments are displayed in the Comments pane, then you can-right click a comment in the pane to open a context menu with the following options:

The context menu for the Comments pane is not enabled if the pane is blank. To add a comment for the sample and enable this context menu, see "The Samples Pane (Run tab) Context Menu" on page 191.

Table 8-6:   Sample tab, Sample Data sub-tab, Comments pane context menu
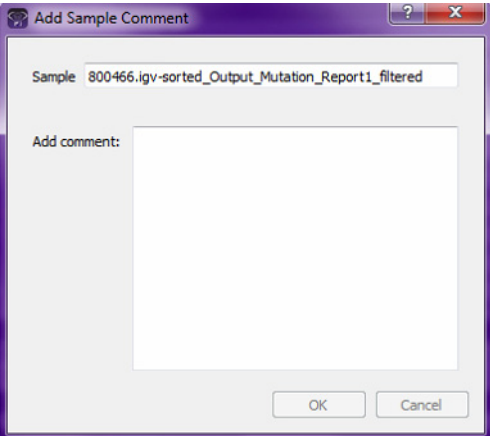
Option	Description
Add Sample Comment	Opens the Add Sample Comment dialog box. You add a comment for the sample in the Add Comment field. *Figure 8-21: Add Sample Comment dialog box* **Note:** If multiple comments have been added for a sample, then these comments are displayed in reverse chronological 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.

Table 8-6:   Sample tab, Sample Data sub-tab, Comments pane context menu (Continued)
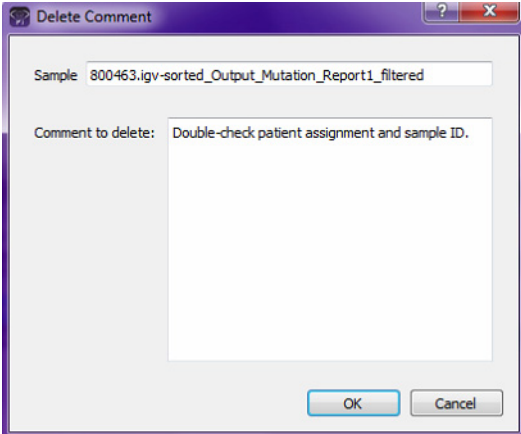
Option	Description
Delete Sample Comment	Opens the Add Delete Comment dialog box. Click OK to delete the comment for the sample and close the dialog box. *Figure 8-22: Delete Comment dialog box*
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Copy	Copies all the information that is displayed for the selected sample to your client's clipboard. You can use standard Paste commands to paste this copied information into a third-party application such as Notepad.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Comments pane. See "To filter the data that is displayed in a tab pane" on page 32.

Table 8-6:   Sample tab, Sample Data sub-tab, Comments pane context menu (Continued)

Option	Description
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the Comments pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Patient pane (Sample Data sub-tab for the Sample tab)

The Patient pane displays information about the patient who is associated with the sample. You can right-click anywhere on the information for the associated patient to open a context menu with the following options:

Table 8-7:   Sample tab, Sample Data sub-tab, Patient pane context menu

Option	Description
Set Patient	Opens the Set Patient dialog box. Select the patient that you are associating with the sample, and then click OK.
	Figure 8-23: Set Patient dialog box
	Note: You also have the option of associating a patient with a sample from the Run tab. See "The Samples Pane (Run tab) Context Menu" on page 191.
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Add Patient External ID to Custom Report	Selects only the External ID for the patient for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
View Details	Opens the Variants sub-tab on the Patient tab for the selected patient. See "Patient tab, Variants sub-tab" on page 158.

Related Patients pane (Sample Data sub-tab for the Sample tab)

The Related Patients pane displays all the relatives of the patient who is associated with the sample. You can right-click anywhere on a related patient record to open a context menu with the following options for working with the patient record:

> *The context menu for the Related Patients pane is not enabled if the pane is blank. To add a patient relationship and enable this context menu, see "To use the context menu for a patient (Patients tab)" on page 141.*

Table 8-8: Sample tab, Sample Data sub-tab, Related Patients 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 137. **Note:** You can also double-click a patient entry to open the Patient tab.
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 one patient at a time" on page 127.
Remove Selected Relationship	Deletes the selected relationship for the patient. To remove multiple relationships, CTRL-click as you select the relationships.
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. See "Setting Read and Write Permissions" on page 326.
Change Ownership	Opens the Change Patient Ownership dialog box, which displays the name of the current owner for the patient, and an option (New Owner) for selecting a new owner for the patient from a list of all the currently available and active users for your Geneticist Assistant instance. You can change the ownership for any patient if you are an Administrator user, the current owner of the patient, or if you have been granted Can Read and Can Write permissions for the patient. *Figure 8-24: Change Patient Ownership dialog box*
Copy	Copies all the information that is displayed for the selected patient to your client's clipboard. You can then use standard Paste commands to paste this copied information into a third-party application such as Notepad.

Table 8-8:   Sample tab, Sample Data sub-tab, Related Patients pane context menu (Continued)

Option	Description
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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:** For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39. **Note:** If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.
\<Saved Pane Configuration Name\> - Saved by \<User Name\>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the Related Patients pane. See "To filter the data that is displayed in a tab pane" on page 32.
\<Filter Template Name\> - Saved by \<User Name\>	Opens the saved Filter template for the Related Patients pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

Compound Variant Interpretations pane

If all the variants that are included in a compound variant interpretation that has been defined in your Geneticist Assistant database are present in the sample, then this variant interpretation is displayed by name in the Compound Variant Interpretations pane. Multiple variant interpretations can be applicable for a given sample. To refresh the pane with the most recent matching interpretations at any time, click Reload. To view the details for all the variants that define a given variant interpretation, double-click the interpretation to open the Compound Variant Interpretation sub-tab. See "Compound Variant Interpretation Sub-Tab" on page 240.

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 includes 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 154.

For detailed information about generating and saving both standard and custom Sample reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.

Compound Variant Interpretation Sub-Tab

If all the variants that are included in a compound variant interpretation that has been defined in your Geneticist Assistant database are present in the sample, then this variant interpretation is displayed by name in the Compound Variant Interpretations pane on the Sample Data sub-tab on the Sample tab. Multiple variant interpretations can be applicable for a given sample. To view the details for all the variants that were used to define a given variant interpretation, double-click the interpretation to open the Compound Variant Interpretation sub-tab.

The Compound Variant Interpretation sub-tab has the exactly the columns as the Sample Variant Data sub-tab and an identical context menu, but instead of displaying all the variants in the sample, it displays only those variants that define the given variant interpretation. (See "Sample Variant Data Sub-Tab" on page 212.)

The Compound Variant Interpretation sub-tab is opened for the Sample tab only after you double-click a variant interpretation in the Compound Variant Interpretations pane on the Sample Data sub-tab on the Sample tab.

Figure 8-25:   Compound Variant Interpretation sub-tab on the Sample tab

Submission Sub-Tab

If the selected sample was part of a submission to the Geneticist Assistant customer portal, then the details of the submission are displayed on the Submission sub-tab. The information that is displayed on the Submission sub-tab and the options that are available such as Download Packing Slip are identical to the information that is displayed for the submission and the options that are available on the Submission tab that is accessed from the Submissions tab.

For detailed information about the Geneticist Assistant customer portal and the Submissions tab, see Chapter 12, "The Geneticist Assistant Customer Portal and Submissions," on page 305.

Figure 8-26: Submission sub-tab on the Sample tab

Quality Metric Report Sub-Tab

If a sample was included in a run for which a quality metric profile was selected, then the Quality Metric Report sub-tab is displayed. The sub-tab details the result metrics for the sample and how they compared to the specified quality metric thresholds.

- If a result metric met or exceeded a Minimum and/or Maximum quality metric threshold limit, then the successful value is not highlighted in the Quality Metric report.

- If a metric failed a Minimum and/or Maximum quality metric threshold, then the failed value is highlighted in red in the Quality Metric report.

- If a metric failed a Warning quality metric threshold limit, then this value is highlighted in yellow in the Quality Metric report.

Figure 8-27:   Sample tab, Quality Metric Report sub-tab
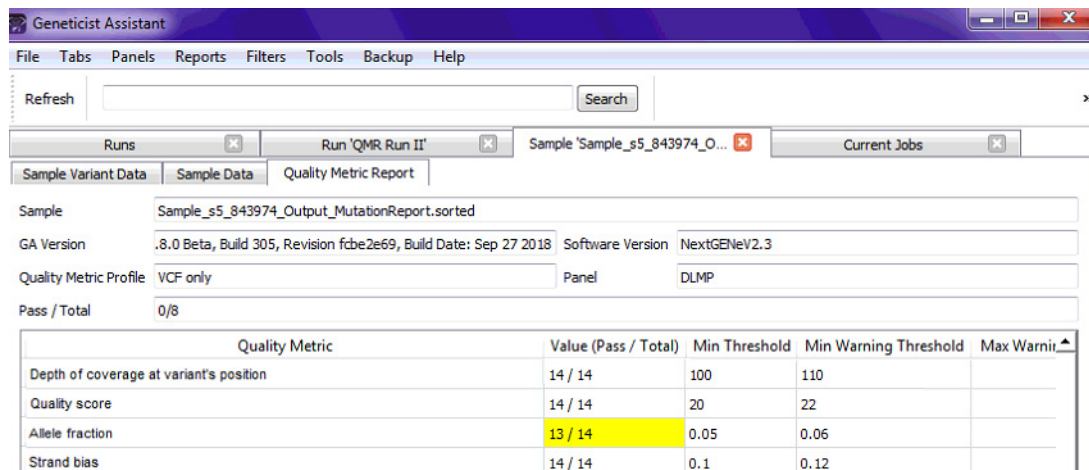
Chapter 9

Geneticist Assistant Data Review - Variant Tab

Many options are available from the Variant tab for reviewing your data in Geneticist Assistant.

This chapter covers the following topics:

- "Overview of the Variant Tab" on page 245.

- "Working with Reference Information Panes on the Variant Tab" on page 256.

Overview of the Variant Tab

The Variant tab displays detailed information for a selected variant. 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 9-1:   *Variant tab*
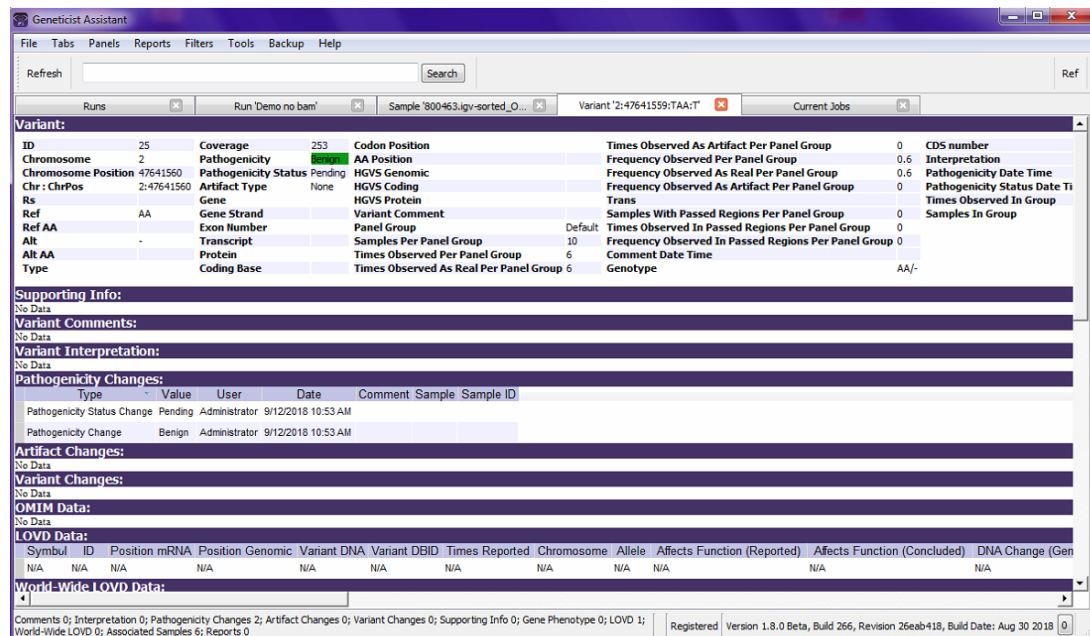
The Variant tab has two types of panes: variant information panes, which display user-specified information for a variant such as its pathogenicity, any comments, its artifact type, and so on, and reference information panes, which display information about the variant that has been culled from a specific reference database such as the dbSNP database. Every pane has a context menu associated with it. Some of the menu commands that are available are common to all the pane context menus, and some are pane-specific. See:

- "Variant information panes" on page 246.

- "Reference information panes" on page 254.

> If any of the reference information panes are not displayed, contact SoftGenetics to download the latest reference files.

Variant information panes

Variant information panes display user-specified information for a variant such as its pathogenicity, any comments, its artifact type, and so on. The following ten variant information panes are displayed on the Variant tab when it opens:

- "Variant pane" on page 247.

- "Supporting Information pane" on page 249.

- "Variant Comments pane" on page 250.

- "Variant Interpretations pane" on page 251.

- "Pathogenicity Changes pane" on page 251.

- "Artifact Changes pane" on page 251.

- "Variant Changes pane" on page 251.

- "Associated Samples pane" on page 252.

- "Variant's Reports pane" on page 254.

- "Gene Phenotypes pane" on page 254.

With the exception of the Variant Interpretations pane, the following commands are displayed on the context menu for every variant information pane:
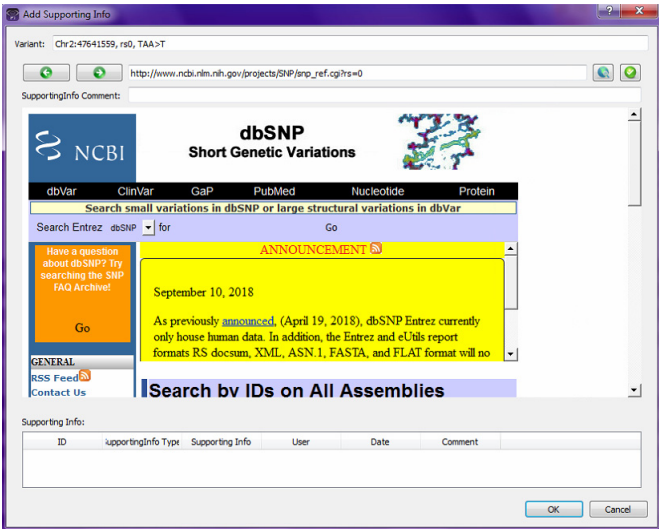
The context menu for a variant information pane is not enabled if the pane is blank.

Option	Description
Add All To Custom Report	Selects all the information that is displayed in the pane for inclusion in a custom report. See Chapter 10, "Geneticist Assistant Reports," on page 265.
Load Columns	
Manually	Opens the Select Columns dialog box in which you can select columns that are to be shown and clear the columns that are to be hidden 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: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 39.
	Note: If you set column display preferences here, then you can 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 69.
Compact	Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.

Option	Description
<Saved Pane Configuration Name> - Saved by <User Name>	Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.
Load Filters	
Manually	Opens the Filters table for the pane. See "To filter the data that is displayed in a tab pane" on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the pane. Limits the display in the pane to only the data (fields) that are defined by the template. See "To filter the data that is displayed in a tab pane" on page 32.

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 Sample Variant Data sub-tab. (See "Sample Variant Data Sub-Tab" on page 212.) You can right-click in the pane to open a context menu that, in addition to the common pane commands, has the following pane-specific commands:
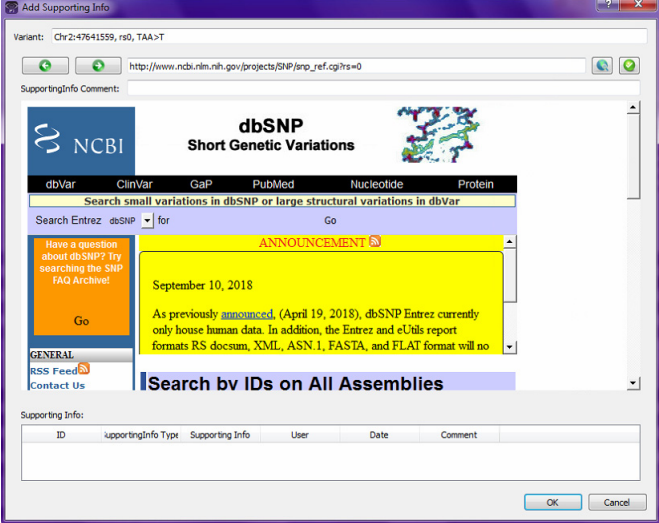
Option	Description
Submit Supporting Hyperlink	Opens the Add Supporting Info dialog box, which, by default, displays the dbSNP web page for the variant. You can use the browser in this dialog box to select this web page as supporting information for the variant, or navigate to a different page. *Figure 9-2: Add Supporting Info dialog box* **Note:** See Step 9 of "To add supporting information for a variant" on page 256.

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, and the Variant Comment field on the Sample Variant Data sub-tab on the Sample tab.

Figure 9-3:   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.

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. (See "To add supporting information for a variant" on page 256.) You can right-click in the pane to open a context menu that, in addition to the common pane commands, has the following pane-specific commands:
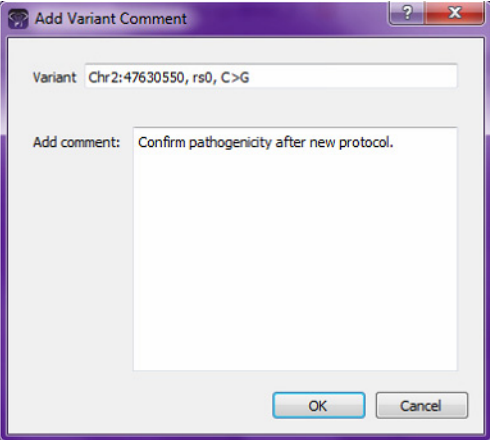
Option	Description
Submit Supporting Hyperlink	Opens the Add Supporting Info dialog box, which, by default, displays the dbSNP web page for the variant. You can use the browser in this dialog box to select this web page as supporting information for the variant, or navigate to a different page. *Figure 9-4: Add Supporting Info dialog box* **Note:** See Step 9 of "To add supporting information for a variant" on page 256.
Delete Supporting Info	Deletes the supporting information. The deleted information is stored in the Geneticist Assistant database log. See "Reviewing the Database Log" on page 50.
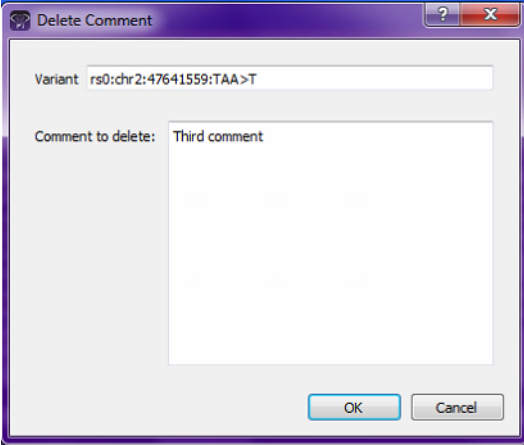
Variant Comments pane

The Variant Comments pane displays all the comments that have been added for the variant in reverse chronological order. If one or more comments are displayed in the Comments pane, then you can right-click in the pane to open a context menu that, in addition to the common pane commands, has the following pane-specific commands:

The context menu for the Comments pane is not enabled if the pane is blank. To add a comment for this variant and enable the pane, see "Variant pane" on page 247.

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, and the Variant Comment field on the Sample Variant Data sub-tab on the Sample tab. *Figure 9-5: 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.

Option	Description
Delete Comment	Opens the Delete Comment dialog box. Click OK to delete the comment for the variant and close the dialog box. *Figure 9-6:* *Delete Comment dialog box*

Variant Interpretations pane

The Variant Interpretations pane is a read-only pane that displays all the single interpretations that have been added for the variant. The Variant Interpretations pane does not have a context menu.

Pathogenicity Changes pane

The Pathogenicity Changes pane displays the history of the changes that have been made to the pathogenicity for the variant in reverse chronological order. When a variant is first added to the Geneticist Assistant database, 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.

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. 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.

Variant Changes pane

The Variant 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
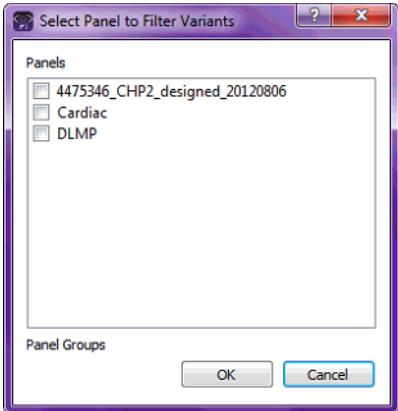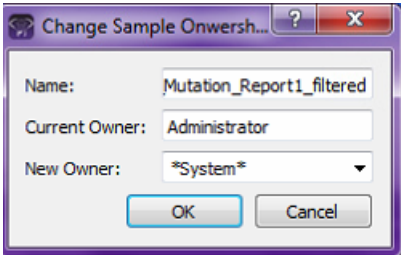
the user who made the change, and the date and the time of the change are recorded and displayed in this pane.

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 that, in addition to the common pane commands, has the following pane-specific commands:

Option	Description
Sample Detail	Opens the Sample tab for the sample. See Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209.
Show Samples Filtered by Panel	Displays all the variants in the selected samples filtered by a selected panel or panels. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel or panels in this dialog box by which to filter the variants for the selected samples. *Figure 9-7:* *Select Panel to Filter Variants dialog box*
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 tab" on page 200.
Panel Statistics	Opens the Panel Statistics tab. The tab has options for displaying specific information based on the panel that was used for a sample's run. See "Reviewing Panel Statistics" on page 319.
Permissions	Opens the Permissions dialog box.You use the options on this dialog box to set or edit the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for selected samples. See "Setting Read and Write Permissions" on page 326.

Option	Description
Change Ownership	Opens the Change Sample Ownership dialog box, which displays the name of the current owner for the selected sample, and an option (New Owner) for selecting a new owner for the sample. You can change the ownership for any sample if you are an Administrator user, the current owner of the sample, or if you have been granted Can Read and Can Write permissions for the sample. *Figure 9-8: Change Sample Ownership dialog box*
External - Contains all the sub-menu options for viewing the sample variants in a tool that is outside Geneticist Assistant.	
View in NextGENe	Requires you to have your NextGENe installation specified in your Geneticist Assistant settings. (See "To configure Geneticist Assistant directories" on page 59.) If selected, then Geneticist Assistant browses for the folder where the VCF file for the sample is located. If the NextGENe project file is found in the same directory as the VCF file, then the NextGENe project is opened in the NextGENe Viewer.
View in IGV	Applicable if there is a corresponding BAM file (and not a NextGENe project file) in the same folder as the VCF file for the sample. Opens the BAM file in the third-party Integrative Genomics Viewer (IGV).
Export - Contains all the sub-menu options for exporting a variant.	
Export to VCF	Exports all the variants in the selected sample or samples (CTRL-click to select multiple samples) to a VCF file. You must specify both the file name and the location in which to save the file.
Save Sample Reports	Applicable for one or more samples. CTRL-click to select multiple samples. Generates an individual sample report for each selected sample. If a single sample is selected, then a default location, report name, and report type (.csv) are provided, but you can change any or all of these values. If multiple samples are selected, then you can select only the location in which to save the report. Each report is named the same as the sample and the report format is .csv, and you cannot change either of these values. **Note:** For more information about standard reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.
Create Batch Variants Report	Generates the Batch Variants report for the selected associated sample or samples. (CTRL-click to select multiple samples.) See "Managing a Batch Variants Report" on page 329.
Create Batch Coverage Report	Generates the Batch Coverage report for the selected associated sample or samples. (CTRL-click to select multiple samples.) See "Generating a Batch Coverage Report" on page 335.

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 includes 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 154.

> *For detailed information about generating and saving both standard and custom Run reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.*

Gene Phenotypes pane

The Gene Phenotypes pane displays all the associated phenotypes for the gene in which the variant was found as pulled from the Human Phenotype Ontology database (https://hpo.jax.org/app/).

Reference information panes

Up to the following ten reference information panes can be displayed when the Variant tab opens:

Database	Description
OMIM Data pane	Displays the information for the variant from the OMIM (Online Mendelian Inheritance in Man) database. **Note:** To view OMIM data, OMIM settings must be configured for your Geneticist Assistant installation. See "Backing up the Geneticist Assistant Database" on page 51.)
LOVD Data pane	Displays the information for the variant from the LOVD (Leiden Open Variation Database) database.
World-Wide LOVD Data pane	Displays information for the variant from the world-wide LOVD (Leiden Open Variation Database) world-wide database.
CIViC Data pane	Displays information for the variant from the CIViC (Clinical Interpretations of Variants in Cancer) database.
gnomAD Data pane	Displays information for the variant from the gnomAD (Genome Aggregation Database) database.
dbSNP (*/00-All.vcf) pane	Displays the information for the variant from the dbSNP database.
Exome Variant Server (*/ESP*.vcf) pane	Displays the information for the variant from the Exome Variant Server database for the NHLBI GO Exome Sequencing Project (ESP).

Database	Description
Exome Aggregation Consortium (ExAC.r*.*.sites.vep.vcf) pane	Displays the information for the variant from the Exome Aggregation Consortium database for selected global sequencing projects.
ClinVar (*/clinvar_00-latest.vcf) pane	Displays the information for the variant from the ClinVar database.
dbNSFP pane	Displays the information for the variant from the dbNSFP database.

You can right-click any information in any of these reference information panes to open a context menu with the following options:

The context menu for a reference information pane is not enabled if the pane is blank.

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 variant" on page 256.
Add Values to Variant Table	Adds values from a selected field in the Reference database to the Variant pane and the Variants pane on the Sample Variant Data sub-tab on the Sample tab. You can also create a custom formula based on multiple fields and add the resulting value to the these same variant panes and tables. See "To add fields from a reference database to variant tables and panes" on page 259 or "To add a custom field to variant tables and panes" on page 261.

Working with Reference Information Panes on the Variant Tab

A variety of options are available for working with the information that is displayed in the various reference information panes on the Variant tab. You can do the following:

- Add supporting information for a variant. See "To add supporting information for a variant" below.

- Add fields from a reference database to variant tables and panes. See "To add fields from a reference database to variant tables and panes" on page 259.

- Add a custom field to variant tables and panes. See "To add a custom field to variant tables and panes" on page 261.

To add supporting information for a variant

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 249.)

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 you are using for supporting information. See "Reference information panes" on page 254.

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 any information 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 9-9:   Supporting Info Submit dialog box
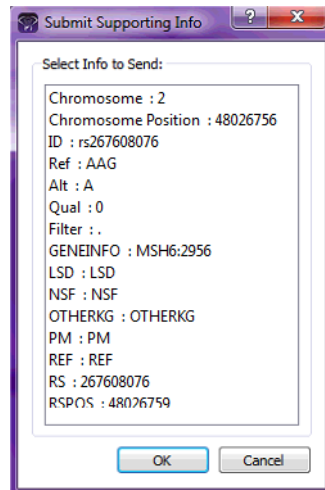
5. Select the information that you are using as supporting information, and then click OK.

The Add Supporting Info dialog box closes. The information is displayed in the Supporting Information pane. See "Supporting Information pane" on page 249.

6. Repeat Step 3 through Step 5 for each piece of reference information that you are adding as supporting information.

7. Optionally, continue to Step 8 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. See Figure 9-10 on page 258.

Figure 9-10:   Add Supporting Info dialog box
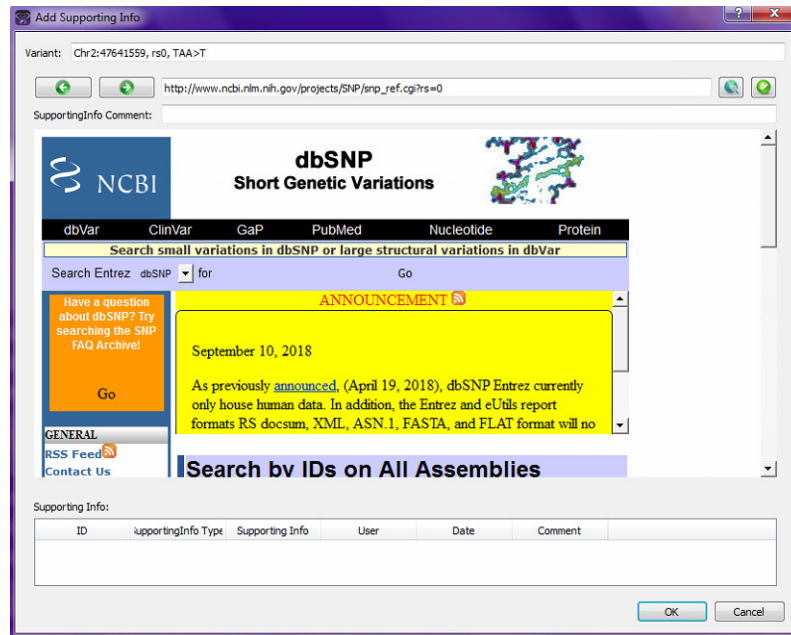
9. Do one of the following:

    *   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 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 249.

To add fields from a reference database to variant tables and panes

You can add fields from a reference database to the Variant pane and the Variants pane on the Sample Variant Data sub-tab on the Sample tab. Fields in the reference database can contain single values, or they can contain multiple values. For example, the Allele Frequency (AF) field in the ExAC Reference database contains a frequency value for the variant nucleotide and a frequency value for the reference nucleotide. Depending on the options that you specify, the field can be added with a just a single value (for example, the value for just the variant nucleotide), or the field can be added with multiple values (the values for both the variant nucleotide and the reference nucleotide).

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. Right-click in the appropriate reference information pane, for example, the ExAC Data pane, and on the context menu that opens, click Add Values to Variant Table.

    The Add Values to Variant Table dialog box opens. The dialog box lists all the fields in the selected Reference database. It also provides a description of the value that is contained in each field.

Figure 9-11:   Add Values to Variant Table dialog box
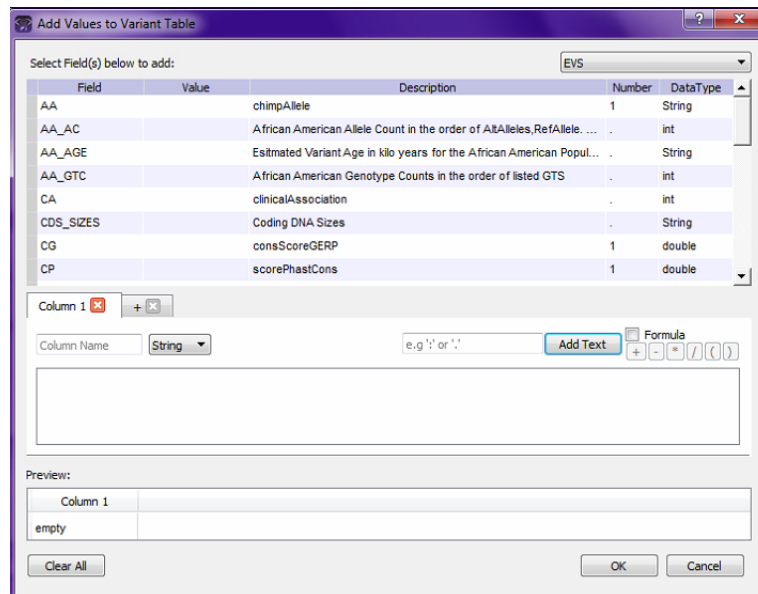
3. Do one of the following to select a field:

    •   Double-click the field.

    •   Right-click the field, and on the context menu that opens, click Add Value(s).

    The Field Configuration dialog box opens. You use the options on the dialog box to specify which values in the selected field are to be added to the various variant panes and tables.

Figure 9-12:   Field Configuration dialog box
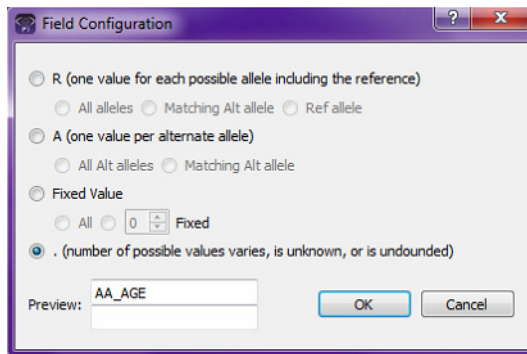
4. Select the values in the field that are to be added to the various panes and tables. The value that is selected by default is determined by how the field is defined in the VCF file for the database.

Value	Description and Options
R (one value for each possible allele including the reference)	The field contains multiple values, with one value for each possible allele, including the reference allele. • All alleles - Add all values for all possible alleles (multiple values) • Matching Alt allele - Add the value only for the matching alternate allele (single value) • Ref allele - Add the value only for the reference allele (single value)
A (one value per alternate allele	The field can contain multiple values, with one value for each alternate allele. • All alt alleles - Add all values for all alternate alleles (multiple values possible if more than one alternate allele exists) • Match Alt allele - Add the value only for the matching alternate allele (single value)
Fixed Value	• All - Add all values (multiple values) • Fixed - Add only the specified number of fixed values, specified value - 1 for the first value, 2 for the second, and so on.
. (number of possible values varies, is unknown, or is unfounded)	Based on different variants, the number of values in the field can vary.

5. Click OK.

    The Field Configuration dialog box closes. The Add Values to Variant Table dialog box remains open.

After you select the values that are to be added, the following happens:

- *A tab that is named the same as the selected field, a Field tab, opens below the list of available fields.*

- *A preview of the selected field and its values is displayed in the Work Area of the dialog box (the blank area below the Field tab).*

- *A preview of the selected field and its values is also displayed in the Preview pane (the bottom pane of the dialog box).*

6. Optionally, to change the name of a Field tab, select the tab to make it the active tab, and then in the Column Name field, enter a new name.

If you add a field in error, then right-click it in the Work Area, and on the context menu that opens, click Remove.

7. Optionally, to add another field, click the (+) tab to open a new blank Field tab, and then repeat Step 3 through Step 6 to add the field to the various panes and tables for the selected variant. Repeat these steps as many times as needed to add all the necessary fields.

8. Click OK on the Add Values to Variant Table dialog box.

    The selected field is added to the various variant tables and panes. The dialog box closes. The Variant tab remains open.

To add a custom field to variant tables and panes

You can combine multiple fields in a reference database to create a custom field, and then add the custom field and its value to the Variant pane and the Variants pane on the Sample Variant Data sub-tab on the Sample tab. When you select a reference database field to use for creating a custom field, if the field contains multiple values, then you can select only a single value from the field. For example, the Allele Frequency (AF) field in the ExAC Reference database contains a frequency value for the variant nucleotide *and* a frequency value for the reference nucleotide. If you select the AF field, then you must select either the frequency value for the variant nucleotide *or* the frequency value for the reference nucleotide. You cannot select both values.

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. Right-click in a Reference pane, and on the context menu that opens, click Add Values to Variant Table.

The Add Values to Variant Table dialog box opens. The dialog box lists all the fields in the selected Reference database. It also provides a description of the value that is contained in each field.

Figure 9-13:   Add Values to Variant Table dialog box
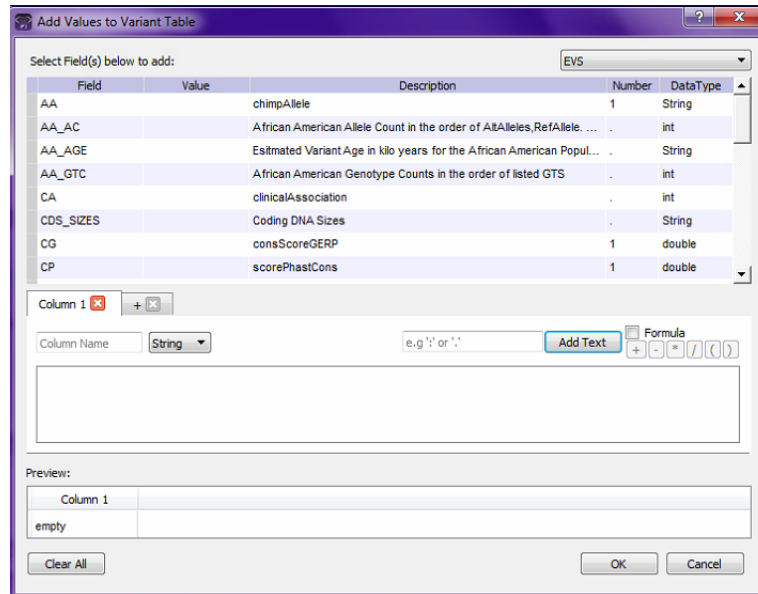
3. Do one of the following to select a field:

- Double-click the field.

- Right-click the field, and on the context menu that opens, click Add Value(s).

The Field Configuration dialog box opens. You use the options on the dialog box to specify the field value that you are using for the custom formula.

Figure 9-14:   Field Configuration dialog box
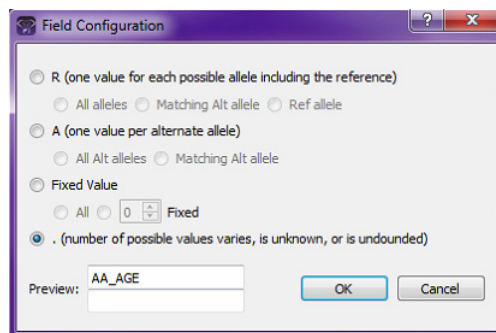
4. Select the value in the field that you are using in the formula for the custom field. The value that is selected by default is determined by how the field is defined in the VCF file for the database.

Value	Description and Options
R (one value for each possible allele including the reference)	The field contains multiple values, with one value for each possible allele, including the reference allele. Therefore, the values that you can select are the following: • Matching Alt allele - Add the value only for the matching alternate allele (single value) • Ref allele - Add the value only for the reference allele (single value)
A (one value per alternate allele	The field can contain multiple values, with one value for each alternate allele. Therefore, the values that you can select are the following: • All alt alleles - Add all values for all alternate alleles (A single value if only one alternate allele exists; however, multiple values possible if more than one alternate allele exists) • Match Alt allele - Add the value only for the matching alternate allele (single value)
Fixed Value	• Fixed - Add only the specified number of fixed values specified value - 1 for the first value, 2 for the second, and so on.
. (number of possible values varies, is unknown, or is unfounded)	Based on different variants, the number of values in the field can vary.

5. Click OK.

After you select the value that you are adding, the following happens:

- *A tab that is named the same as the selected field, a Field tab, opens below the list of available fields.*

- *A preview of the selected field and its value is displayed in the Work area of the dialog box (the blank area below the Field tab).*

- *A preview of the selected field and its value is also displayed in the Preview pane (the bottom pane of the dialog box).*

6. Do one of the following:

- Click in the Add Text field, enter the needed text (for example, if you are creating a field to show a ratio, you might select a forward slash), and then click Add Text.

  The text is displayed on the Field tab and in the Work Area and the Preview pane.

- To create a formula, select Formula, and then select the operator for the formula. Parentheses are also available for setting the order of operations in the formula.

  As you create the formula, the following happens:

- A preview of the formula, including the selected fields, their values, and the operators, is displayed on the Field tab and in the Work area of the dialog box (the blank area below the tabs).

- The Preview pane displays the current value for the formula.

If you add a field, a text separator, or an operator in error, then right-click it in the Work Area, and on the context menu that opens, click Remove.

7. Repeat Step 3 through Step 6 as needed to produce the final custom field.

   As you select multiple fields for the custom field, the display on the Field tab and in the Work Area and the Preview pane are dynamically updated.

8. Optionally, to add another custom field, click the (+) tab to open a new blank tab, and then repeat Step 3 through Step 7. Repeat these steps as many times as needed to add all the necessary custom fields.

9. Optionally, to change the name of the Field tab, select the tab to make it the active tab, and then in the Column Name field, enter a new name for the tab.

10. Click OK.

    The Field Configuration dialog box closes. The Add Values to Variant Table dialog box remains open.

11. Click OK on the Add Values to Variant Table dialog box.

    The custom fields are added to the various variant tables and panes. The dialog box closes. The Variant tab remains open.

Chapter 10
Geneticist Assistant Reports

As discussed in the various data review chapters in this manual, 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. Two options are available for generating an output file of the data: you can generate a standard report, or you can select some or all of the data for inclusion in a custom report. You can also use the Report Designer to create a *Sample PDF Report template*. A Sample PDF Report template not only specifies the data that a Sample PDF report is to contain, but also the layout of the report.

This chapter covers the following topics:

- "Generating a Standard Report" on page 267.

- "Generating a Custom Report" on page 271.

- "Designing a Sample PDF Report Template" on page 274.

- "Generating a Sample PDF Report" on page 286.

Generating a Standard Report

To generate an output file of the data after a run is successfully completed, you generate a *standard* report. A standard report includes all the data that is displayed in a selected pane or panes 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 and/or the Run's Status Changes pane on the tab.

- Coverage Regions report—You generate the Coverage Regions report from the Coverage Regions tab. The report includes all the information that is displayed on the 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:

   - Chapter 7, "Geneticist Assistant Data Review - Run Tab," on page 187.

   - "Coverage Regions tab" on page 200.

   - The Panel Statistics tab. See "Reviewing Panel Statistics" on page 319.

   - Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209.

   - Chapter 9, "Geneticist Assistant Data Review - Variant Tab," on page 243.

   To generate an Action List report, you must first create the Action List to open the Action List tab. See "Creating an Action List" on page 338.

2. Do one of the following:

Table 10-1: Available standard reports

Report to be Generated	Action
Run	On the Geneticist Assistant main menu, click Reports > Run Report.
Coverage Regions	On the Geneticist Assistant main menu, click Reports > Coverage Regions 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 10-1:   Run Report dialog box
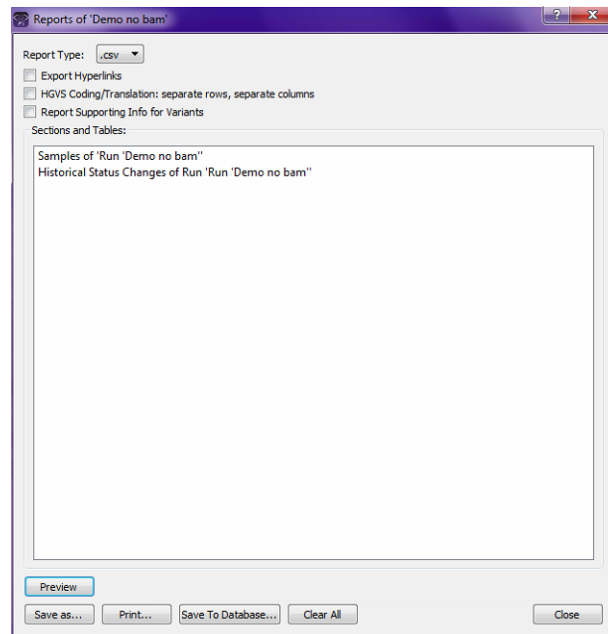
3. Optionally, to remove a sub-tab or pane from the report, right-click the row for the sub-tab or pane, and then click "Remove Selected Row" to remove the data from the indicated pane or sub-tab from inclusion in the report.
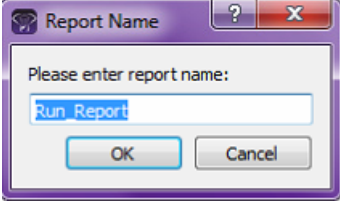
Only the data columns that are shown in the selected sub-tabs or panes at the time that the report is generated are included in the report. Hidden data columns are not included. See "To show/hide columns in a tab pane" on page 39.

4. Leave the report type set to the default value of ..csv, or select .xml.

5. Optionally, to include active hyperlinks in the saved reports, select Export Hyperlinks.

6. Optionally, for reports that contain variants, such as the Sample report, the Variant report, or the Action List report, select one or both of the following:

   • HGVS Coding/Translation: separate row, separate columns: If multiple transcripts are shown for the HGVS Coding or HGVS Protein for a variant, then select this option to create a separate row for each transcript and two separate columns—one for the transcript ID and one for the actual call—within each row.

   • Report Supporting Info For Variants: If supporting information has been added for any of the variants for which you are generating the report, then select this option to include this supporting information in a separate Supporting Information section in the report.

7. Do any or all of the following:

Table 10-2: 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.

Table 10-2: Standard report options (Continued)

Option	Description
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 is displayed for the report. For example, if you generate a standard Sample report, then the default report name is always the sample name. *Figure 10-2: 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.
Note: If filters were applied to the data for which you are generating the report, then these filters are explicitly displayed in a separate Filters Applied section in the report.	

8.   Click Close to close the Report dialog box.

Generating a Custom Report

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 or tabs.

> *When you generate custom report, you specify the data that you are including in the report, but the report is generated according to a standard layout and format. You also have the option of using the Report Designer to generate a custom PDF report for which you not only select the data, but also specify the layout and format. See "Designing a Sample PDF Report Template" on page 274.*

To generate a custom report

1. Open the appropriate tab. See:

   - Chapter 7, "Geneticist Assistant Data Review - Run Tab," on page 187.

   - "Coverage Regions tab" on page 200.

   - The Panel Statistics tab: See "Reviewing Panel Statistics" on page 319.

   - Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209.

   - Chapter 9, "Geneticist Assistant Data Review - Variant Tab," on page 243.

> *To generate an Action List report, you must first create the Action List to open the Action List tab. See "Creating an Action List" on page 338.*

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 CTRL-click 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 "To show/hide columns in a tab pane" on page 39.*

3. On the Geneticist Assistant main menu, click Reports > Custom Report.

   The Custom Report dialog box opens. All the data that you are including in the report is displayed in the Sections and Tables pane of the dialog box.

Figure 10-3:   Custom Report dialog box
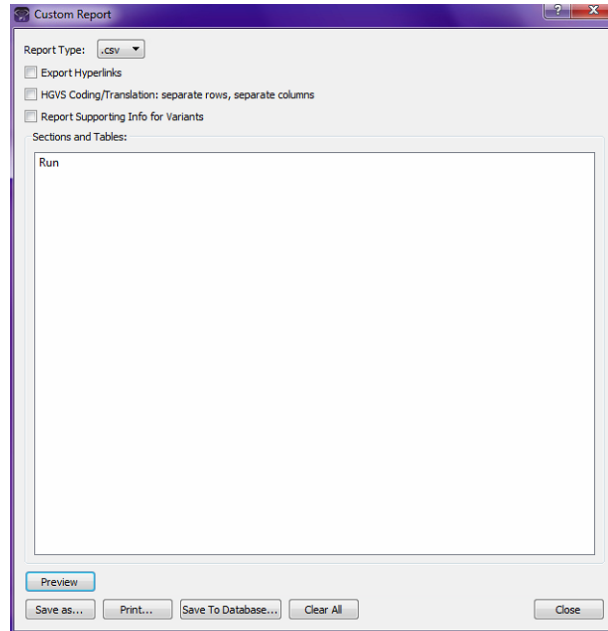
4. Optionally, if data has been selected in error for the custom report, or you want to remove some selected data before you generate the report, right-click the entry in the Sections and Tables pane, and on the context menu that opens, click Remove Selected Row.
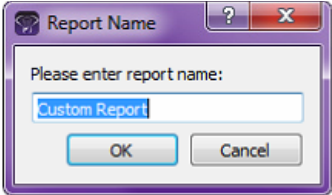
   If you previously generated a custom report, and you did not click Clear All before you clicked Cancel, or closed the report dialog box, then any new information that you select on any tab for inclusion in a custom report is simply appended to this existing report.

5. Leave the report type set to the default value of ..csv, or select .xml.

6. Optionally, to include active hyperlinks in the saved reports, select Export Hyperlinks.

7. Optionally, for reports that contain variants, such as the Sample report, the Variant report, or the Action List report, select one or both of the following:

   • HGVS Coding/Translation: separate row, separate columns: If multiple transcripts are shown for the HGVS Coding or HGVS Protein for a variant, then select this option to create a separate row for each transcript and two separate columns—one for the transcript ID and one for the actual call—within each row.

   • Report Supporting Info For Variants: If supporting information has been added for any of the variants for which you are generating the report, then select this option to

include this supporting information in a separate Supporting Information section in the report.

8. Do any or all of the following:

Table 10-3: 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 10-4: 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.

9. Click Clear All.

This ensures that information that is selected for a new custom report is not simply appended to an existing report.

10. Click Close to close the Report dialog box.

Designing a Sample PDF Report Template

A *Sample PDF report template* specifies not only the data that a Sample PDF report is to contain, but also the layout of the report. The PDF Report Designer provides a user-friendly interface for building and saving a robust custom report template.You can then select this template to generate the corresponding Sample PDF report for specific data in your Geneticist Assistant database. The PDF Report Designer option is available from the Reports menu only from an open Sample tab: Open a Sample tab, and then on the Geneticist Assistant main menu, select Reports > PDF Report Designer.

The PDF Report Designer user interface

This section is designed to provide an introduction to the basics of using the PDF Report Designer to design a Sample PDF Report template. For assistance with advanced functions and/or designing a report template, contact tech_support@softgenetics.com.

When you are designing a Sample PDF report template, you have two parts to the design. You must specify the filters and sort order for the table of variants that is contained in a Sample PDF report that is based on the template, and then you must design the report template. You specify the filters and sort order in the PDF Report Designer Data Sources dialog box, and you design the report template in the PDF Report Designer dialog box. The PDF Report Designer Data Sources dialog box opens first after you select the PDF Report Designer option on the Reports menu. It displays a placeholder for the table of variants with the following default values:

- Filter - None. The table of variants is not filtered in the Sample PDF report.

- First sort and Sort Order - The table of variants is sorted first by Chromosome and Chromosome Position (Chr:ChrPos) in ascending order (Asc).

- Second Sort and Sort Order - The table of variants is sorted second by Pathogenicity in ascending order.

Figure 10-5:   PDF Report Designer Data Sources dialog box
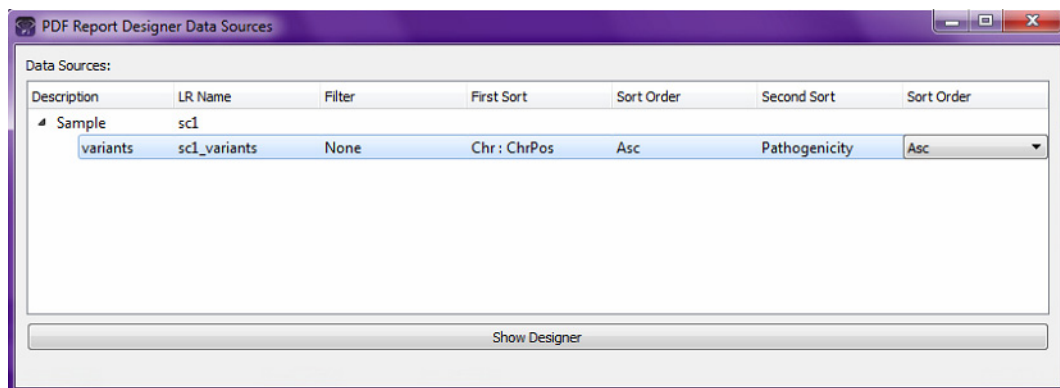
You can leave the default values as-is, or you can change any or all of the values. To change the Filter option, a Sort option, or a Sort Order option, double-click in the appropriate field to open a dropdown list, and then select a new value.

- Filter - Select from a list of Variant Filters (displayed by name) that you have added to the Geneticist Assistant or that other users have added to the database and made available to all users.

- Sort - The available sort options are pulled from the variant annotation columns of the Sample Variant Data sub-tab columns.

- Sort Order - Two values are available, ascending (Asc) and descending (Desc).

After you have set the values for the table of variants, then at the bottom of the PDF Report Designer Data Sources dialog box, click Show Designer to open the PDF Report Designer. When the PDF Report Designer opens, all panes other than the Data Browser pane and Script pane are blank. The Report Structure pane displays a placeholder for adding a new object to the first page of the report template.

Figure 10-6:   PDF Report Designer

Table 10-1: PDF Report Designer user interface

Option	Description
1	Object Inspector toolbar. Contains options for adding objects, such as a text field, to a report page. Hold your mouse pointer over an icon on the toolbar to open a tooltip that explains the icons' function. See "Object Inspector toolbar" on page 276.
2	Object toolbar. Contains options for working with objects on a report page, such as aligning objects or snapping objects to the grid. Hold your mouse pointer over an icon on the toolbar to open a tooltip that explains the icons' function. See "To add and arrange Data band items" on page 278.

Table 10-1: PDF Report Designer user interface

Option	Description
3	Report Structure pane. Lists all the bands and objects that are displayed on a currently opened page of a Sample PDF Report template.
4	Work Area. You build a Sample PDF Report template in the Work Area. See "Sample PDF Report template layout" on page 277.

Object Inspector toolbar

The Object Inspector toolbar contains options for adding objects such as a text field to a report template page. Hold your mouse pointer over an icon on the toolbar to open a tooltip that explains the icon's function. Table 10-2 below provides an overview of the some of the relevant icons on the toolbar.

Table 10-2: Object Inspector toolbar

Icon	Description
	Report bands icon - Opens a Bands dropdown list which displays all the different types of bands that you can add to a Sample PDF Report template.
	Text Item icon - Use this icon to add a text field to a Data band.
	Chart Item icon - Use this icon to add a chart to a Data band.
	Image Item icon - Use this icon to add an image to a Data band. Allowed image formats are .png, .bmp, and .jpeg/.jpg.
	Delete Item icon - Use this icon to delete a selected item or image placeholder from a Data band. **Note:** To delete an image from a Data band, but leave the image placeholder, you must use the Delete Image icon in the Object Inspector. See "To add an image to a Data band" on page 281.

Sample PDF Report template layout

A Sample PDF Report template is built around a single data table or multiple data tables referred to as *Data bands*. Any Data band can have an associated Data header band and/or a Data footer band. Other optional components of a custom report template include the following: Report Header, Report Footer, Page Header, Page Footer, SubDetail, SubDetailHeader, SubDetail Footer, GroupHeader, and GroupFooter.

Figure 10-7: Sample PDF Report template in the Work Area of the Report Designer
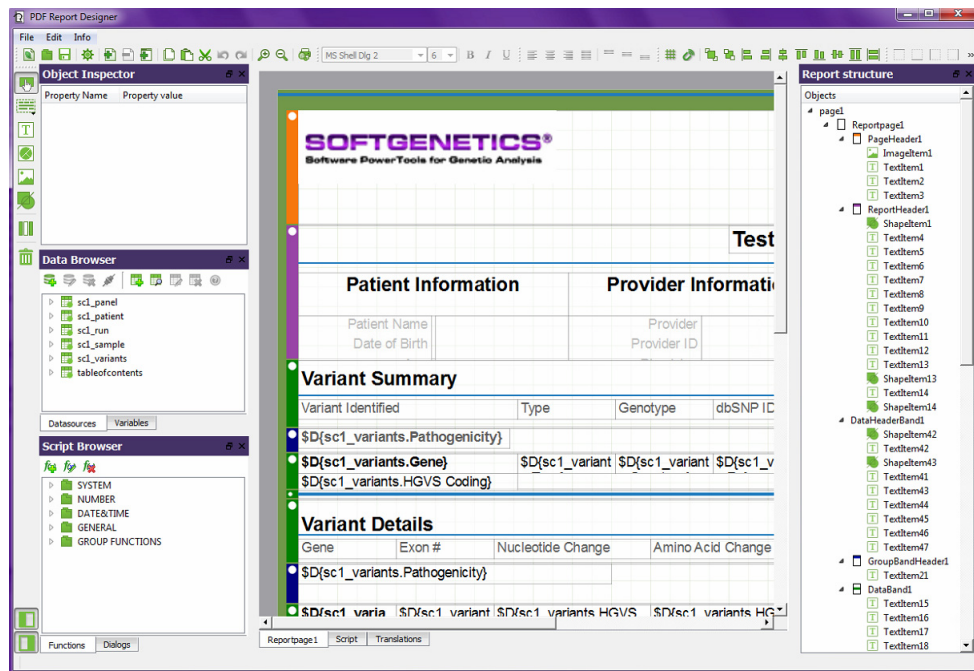
After you add the Data bands for a custom report template, you then specify the database and or/ text fields that are to be included in each band. You can also add one or more images to a band. You can also arrange the items in a Data band as you are adding them to the band or after you have added them to the band. For example, you can align all text fields vertically. After you have designed the template, you can then save the report template to the Geneticist Assistant database. See any of the following for a high-level description of building a Sample PDF Report template:

- "To add Data bands to a Sample PDF Report template" below.

- "To add and arrange Data band items" on page 278.

- "To save a new Sample PDF Report template" on page 283.

To add Data bands to a Sample PDF Report template

You can add all the required Data bands for a report template, and then add the required fields and/or images to each Data band, or you can add one Data band at a time and then the fields and/or images for the band. Before you can add a Data band header or Data band footer, you *must* have the Data band added to the template and you must select it.

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

2. On the Geneticist Assistant main menu, select Reports > Report Designer.

   The PDF Report Designer Data Sources dialog box opens.

3. Leave the default data source values as-is, or edit or all of the values as appropriate, and then at the bottom of the dialog box, click Show Designer.

   For information about the PDF Report Designer Data Sources dialog box, and the values that you can set in this dialog box, see "The PDF Report Designer user interface" on page 274.

   The PDF Report Designer opens. When the Report Designer opens, it is blank. The Work Area displays the first page (Page 1) for the report.

4. For each Data band that you are adding to the report template, do the following:

   a. If you are adding a Data header or Data footer to a Data band, then click the Data band to select it.

      The band is highlighted in red, with "grab" handles (red circles) displayed on its outline.

   b. On the Object Inspector toolbar, click the Report bands icon.

      A Bands menu opens.

   c. On the Band menu that opens, click the appropriate band type, for example, Data.

      The band is added to Page 1 of the report in the Work Area, and it is automatically selected. The band is labeled as <band type(n)>, for example, ReportHeader1.

Figure 10-8: Data band added for a Sample PDF Report template
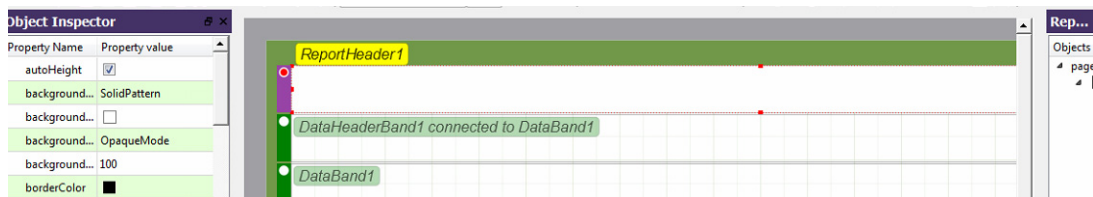
5. Continue to "To add and arrange Data band items" below.

To add and arrange Data band items

After you add a Data band to a Sample PDF Report template, you can add a variety of different items to the band, such as a database field, a text field, and/or an image. Options are also available for arranging the items as you add them to the Data band and/or after you add them to the Data band, for example, aligning items to the grid, or horizontally aligning all objects in the Data band. The following sections provide a high-level overview of adding and arranging these various Data band items.

To add a database field to a Data band

The Data Browser pane displays all the data sources (fields) that are stored in the Geneticist Assistant database that you can include in a Sample PDF Report template. The data sources are organized by data type (tabs), with each data source further organized based on the information that is available for the data type. Five data types are available: Panel, Patient, Run, Sample, and Variants. To add a database field to a report template, do the following:

In the Data Browser pane, expand the data type to see its available fields, and then drag and drop a selected field on to a Data band.

Figure 10-9: Adding a database field to a Data band
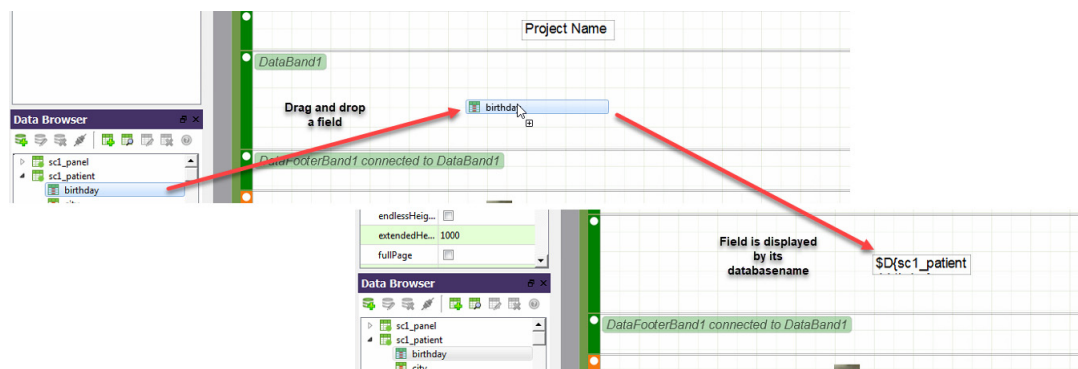
You can add multiple database fields to the Data band, as well as text fields and/or images. You can also arrange the fields as you add them or after you add them. If you are done designing the report template, then you can save the template. See:

- "To add a text field to a Data band" below.

- "To add an image to a Data band" on page 281.

- "To arrange items in a Data band" on page 282.

- "To save a new Sample PDF Report template" on page 283.

To add a text field to a Data band

A Text item is a text field in which you can enter free text. To add a text field to a report template, to the following:

1. On the Object toolbar, click the Text Item icon, and then move your mouse pointer to the appropriate location in the Data band.

   A blank Text item placeholder is displayed.

Figure 10-10: Blank text item placeholder
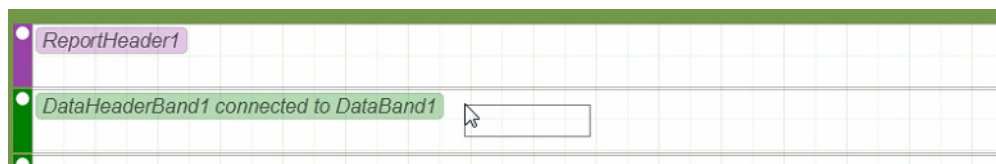
2. Click to place the text field in the selected location.

   The text field is automatically selected, and resizing handles are displayed on its outline.

Figure 10-11: Placed text field

3. Double-click the selected text field.

   The Text Item Editor dialog box opens.

Figure 10-12: Text Item Editor dialog box
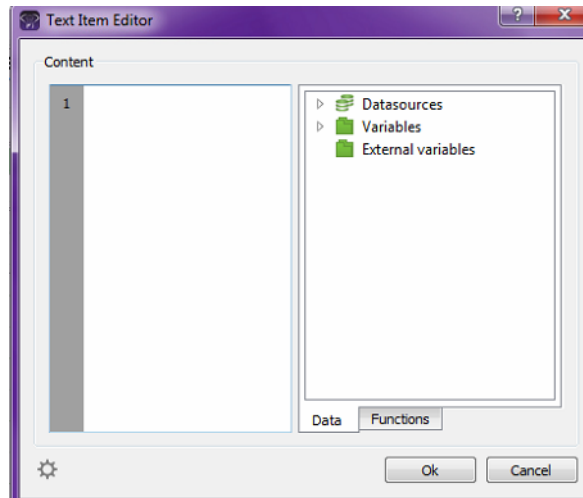
4. In the blank Content pane, enter the text for the field, and then click OK.

   The Text Item Editor dialog box closes. The content that you entered for the text field is displayed in the field on the report template.

Figure 10-13: Text in text field
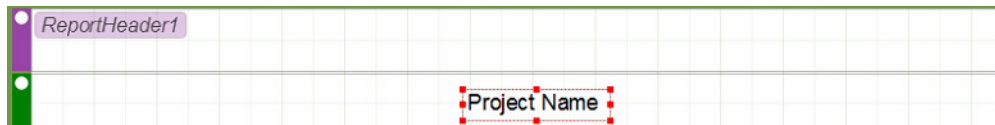
You can add multiple text fields to the Data band, as well as database fields and/or images. You can also arrange the fields as you add them or after you add them. If you are done designing the report template, then you can save the template. See:

- "To add a database field to a Data band" on page 279.

- "To add an image to a Data band" on page 281.

- "To arrange items in a Data band" on page 282.

- "To save a new Sample PDF Report template" on page 283.

To add an image to a Data band

In addition to database fields and free text fields, you can add images in a PNG (.png) format, BMP (.bmp) format or JPEG/JPG (.jpeg/.jpg), to a report band.

1. On the Objects toolbar. click the Image Item icon, and then move your mouse pointer to the appropriate location in the Data band.

   An Image item placeholder is displayed.

2. Click to place the image placeholder in the selected location.

   The image placeholder is automatically selected, and resizing handles are displayed on its outline.

Figure 10-14: Image placeholder in a Data band
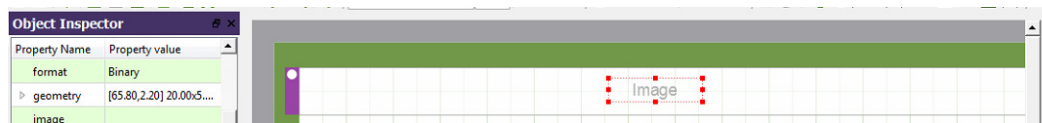
3. In the Object Inspector pane, scroll to the image field, and then click in the field.

   Two icons are displayed in the image field: The left icon is the Open Image icon and the right icon is the Delete Image icon.

Figure 10-15: image field icons in the Object Inspector
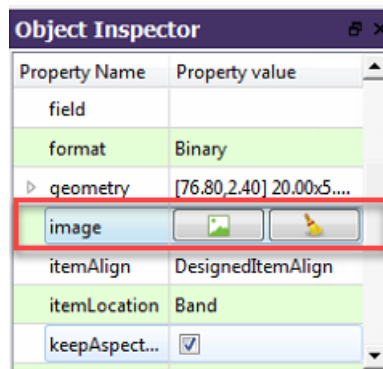
4. Click the Open Image icon, and then in the Open dialog box, browse to and select the image that you are adding.

   The Open Image dialog box closes, and the selected image replaces the "Image" placeholder text.

If you add an incorrect image, or want to remove an image from a Data band, then click the image to select it, and in the image field in the Object Inspector pane, click the Delete Image icon. To delete the image placeholder, select it, and then on the Object toolbar, click the Delete Item icon.

You can add multiple images to the Data band, as well as add database fields and/or text fields. You can also arrange the fields as you add them or after you add them. If you are done designing the report template, then you can save the template. See:

- "To add a database field to a Data band" on page 279.

- "To add a text field to a Data band" on page 279.

- "To arrange items in a Data band" below.

- "To save a new Sample PDF Report template" on page 283.

To arrange items in a Data band

The Object toolbar contains options for working with objects on a report template page, such as aligning objects or snapping objects to the grid. You can hold your mouse pointer over an icon on the toolbar to open a tooltip that explains the icons' function. Table 10-3 below describes the icons on this toolbar.

Table 10-3: Object toolbar icons

Icon	Description
▦	Use Grid icon - Displays a grid in each Data band with dimensions that are based on the size of the Data band. Limits the size and position of items in the Data band to the grid's dimensions.
⬭	Use Magnet icon - Aligns objects in the Data band to each other based on the height, width, and vertical and horizontal position of a selected object. The first item that you select in a Data band with this icon selected is the object to which all other objects are aligned.
⬚	Bring to top icon - If two or more items overlap in a Data band, then select the item that you are bringing to the top, and then click this icon.
⬚	Send to back icon - If two or more items overlap in a Data band, then select the item that you are sending to the back, and then click this icon.
▤	Align to left icon - CTRL-click to select multiple items, and then click this icon to align the left edges of all selected items to the left edge of the first selected item.
⬌	Align to right icon - CTRL-click to select multiple items, and then click this icon to align the right edges of all selected items to the right edge of the first selected item.
⬌	Align to vertical center - CTRL-click to select multiple items, and then click this icon to align the vertical centers of all selected items to the vertical center of the first selected item.
▯▯	Align to top icon - CTRL-click to select multiple items, and then click this icon to align the top edges of all selected items to the top edge of the first selected item.
▯▯	Align to bottom icon - CTRL-click to select multiple items, and then click this icon to align the bottom edges of all selected items to the bottom edge of the first selected item.

Table 10-3: Object toolbar icons (Continued)

Icon	Description
⬦	Align to horizontal center - CTRL-click to select multiple items, and then click this icon to align the horizontal centers of all selected items to the horizontal center of the first selected item.
⬦	Set same height icon - CTRL-click to select multiple items, and then click this icon to set the heights of all selected items to the height of the first selected item.
⬦	Set same width icon - CTRL-click to select multiple items, and then click this icon to set the widths of all selected items to the width of the first selected item.

After you have arranged the items in a report template, you can continue adding other items as needed, or if you are done designing the report template, then you can save the template. See:

- "To add a database field to a Data band" on page 279.

- "To add a text field to a Data band" on page 279.

- "To add an image to a Data band" on page 281.

- "To save a new Sample PDF Report template" below.

To save a new Sample PDF Report template

After you have designed the Sample PDF Report template, you can then save the report template to the Geneticist Assistant database as new template.

For other options for working with a report template, such as editing an existing report template, see "Other PDF Sample Report Template options" on page 284.

1. On the PDF Report Designer dialog box main menu, click File > Save Report.

   The Save Report Template dialog box opens. The dialog box displays all the saved Sample PDF Report templates by name.

Figure 10-16: Save Report Template dialog box
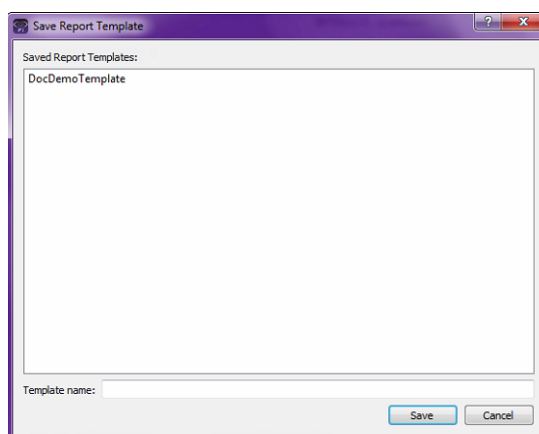
2. In the Template Name field, enter a name for the report template.

3. Click Save.

   The Save Report Template dialog box closes. The report template is saved to the Geneticist Assistant database. The PDF Report Designer dialog box remains open.

4. In the upper right corner of the PDF Report Designer dialog box, click the Close (x) button to close the dialog box.

   The PDF Report Designer Data Source dialog box remains open.

5. In the upper right corner of the PDF Report Designer Data Sources dialog box, click the Close (x) button to close the dialog box, and return to the Geneticist Assistant main window.

Other PDF Sample Report Template options

In addition to saving a PDF Sample Report template as a new template, the following options are available from the File menu on the PDF Report Designer dialog box:

•   New Report - Clears all items from the Work Area and leaves the Work Area blank in preparation for designing a new Sample PDF Report template.

•   Load Report - Opens the Load Report Template dialog box. The dialog box displays all the saved Sample PDF Report templates by name. You can select a report template from this list of saved templates, and then click Load to close the dialog box and load the report in the PDF Report Designer Report dialog box.

Figure 10-17: Load Report Template dialog box
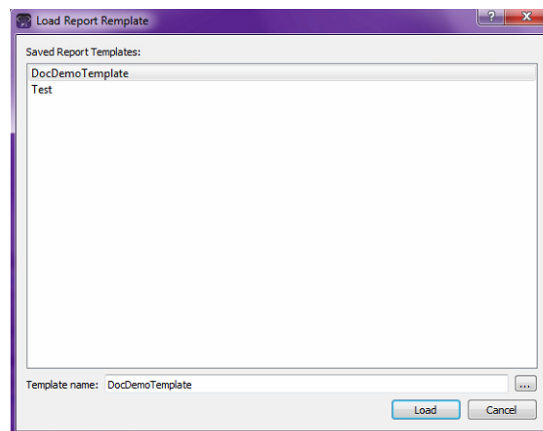
•   Save Report - Saves the currently loaded Sample PDF Report template without changing its name. Any changes that you make and save overwrite the existing report template.

•   Save as - Opens the Save Report Template dialog box. The current name for the report template is displayed in the Template name field. You can enter a new template name to save the report template as a new template.

Save As is a useful option for creating a new report template by editing an existing template, and then saving the template with a new name. You do not have to create an entirely new report template.

- Render report - Generates an onscreen preview of the Sample PDF report. The report demonstrates the format and the contents of any Sample PDF report that will be generated based on this report template. A toolbar is displayed at the top of the Preview dialog box with icons for working with the preview, such as zooming in or zooming out the preview. Hold your mouse pointer over an icon on the toolbar to open a tooltip that explains the icons' function.

Figure 10-18: Sample PDF Report Template preview
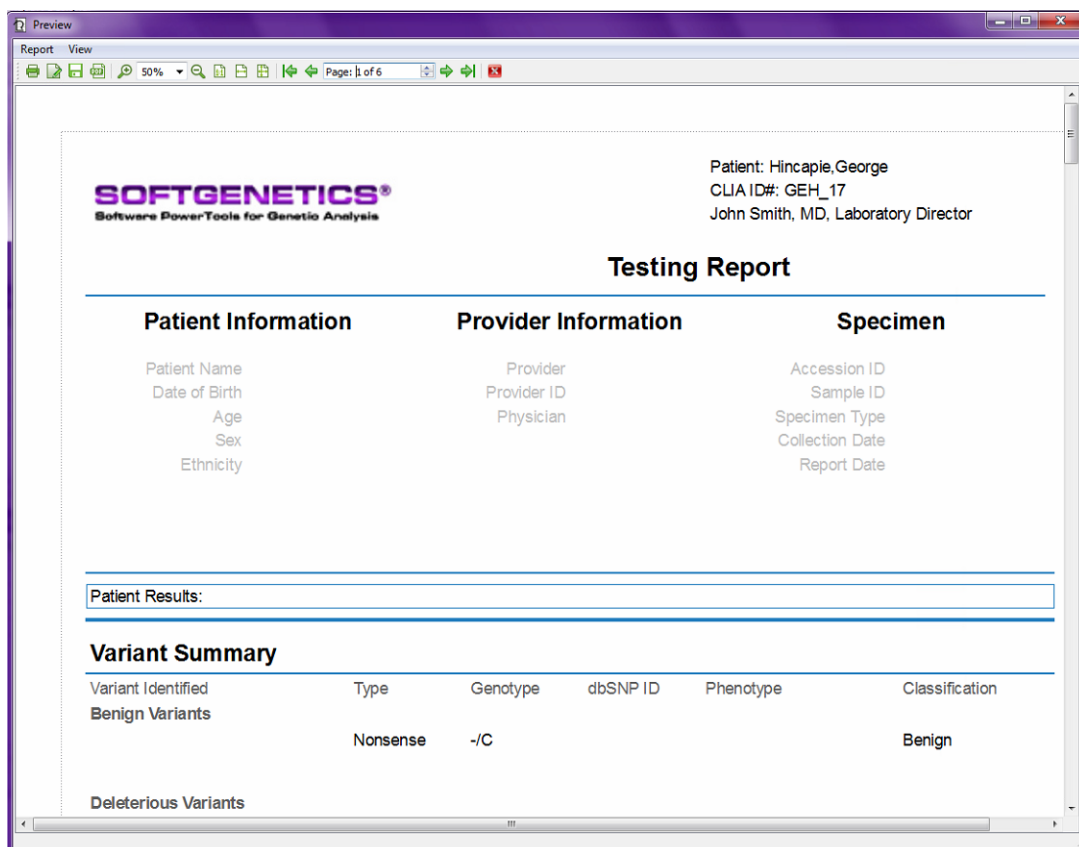
Generating a Sample PDF Report

A Sample PDF Report details specific information about the variants that are found in the sample based on a Sample PDF Report template. You can generate a Sample PDF report from the following locations:

- Sample tab. (From the Reports menu with the Sample tab open.)

- Runs tab context menu.

- Run tab context menu.

When you generate the report, the report is saved in as a PDF, and you cannot change this format. You must specify a name and location in which to save the report.

To generate a Sample PDF report

1. Open the Sample tab for the appropriate sample. See "Overview of the Sample Tab" on page 211.

2. On the Geneticist Assistant main menu, click Reports > Sample PDF Report.

   A sub-menu opens that displays all the Sample PDF Report templates that have been saved to the Geneticist Assistant database. See "Designing a Sample PDF Report Template" on page 274.

3. Select the Sample PDF Report template.

   The Save Sample PDF Report dialog box opens.

4. Select a location in which to save the report, enter a file name, and then click Save.

   The Save Sample PDF Report dialog box closes and the report is generated and saved in the selected location.

Chapter 11
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 converting a VCF file to a BED file, a tool for managing pathogenicity changes, a tool for specifying pathogenicities for specific variants in a panel, a tool for finding samples in your Geneticist Assistant that contain specific variants, and a tool for combining BAM files into a single sample.

This chapter covers the following topics:

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 > Build BED file.

   The BED File Builder dialog box opens.

   Figure 11-1: BED File Builder dialog box
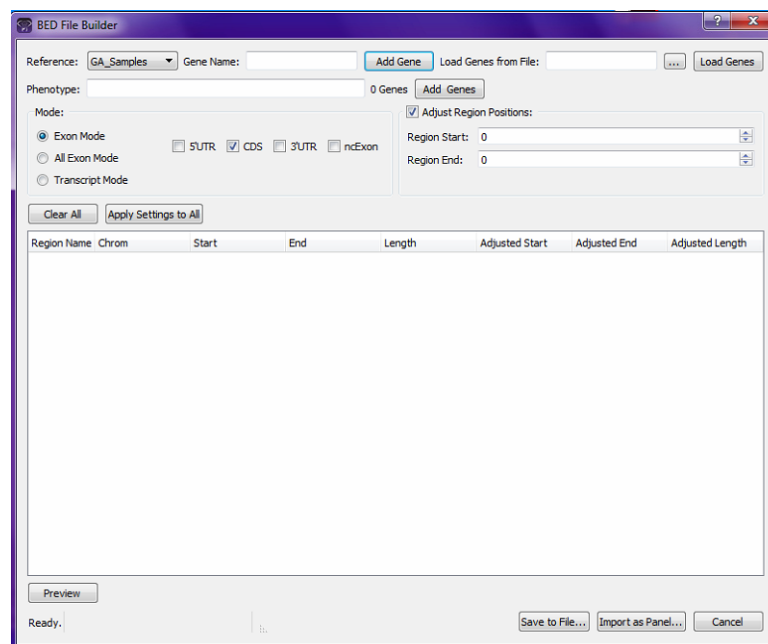
2. Do one of the following:

   - 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 search is not case-sensitive, but the search string must begin with the gene name. For example, a search string of "as" returns ASH1L-AS1 but a search string of "as1" does not.

- 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. If you are building a BED file for genes that are associated with a specific phenotype, then do the following; otherwise, go to Step 4.

In the Phenotype field, enter a search string for the phenotype. As you enter the search string, a dropdown list opens. The list is dynamically updated with the phenotypes that are found in the Human Phenotype Ontology database that match the search string.

> *The search string is not case-sensitive, and the string can be found anywhere in the search results. For example, a search string of "thyroid" returns phenotypes of **thyroid** adenoma and secondary hyper**thyroid**ism.*

4. 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 specified exons from all the selected transcripts in the BED file. You can include only the coding regions in the BED file, only the untranslated regions, or both. Introns are *not* included in Exon mode.

5. Do one of the following:

- For Exon mode and All Exon mode—Select a 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 11-2: BED File Builder tool, Exon mode
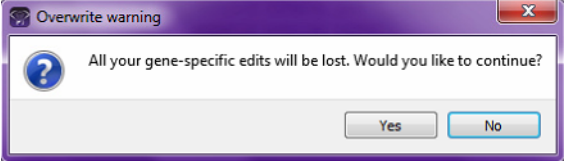
Select an entire transcript, or click the Expand icon.

Specify the coding region and/or untranslated regions.
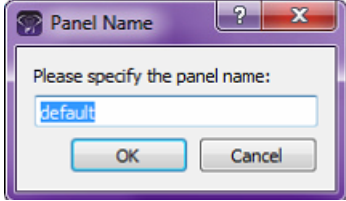
- For Transcript mode—Select a transcript.

6. If you are building a BED file for genes that are associated with a specify phenotype, then click Add Gene; otherwise, go to Step 7.

The appropriate regions are added for the BED file based on the selected Mode. See Step 4.

7. Optionally, do any or all of the following:

Option	Steps
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. **Note:** 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 and the Start/End Region values) to all genes, including any previously added genes	1. Click Apply 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. *Figure 11-3: Overwrite warning message* 2. Click Yes to close the message and overwrite all the current gene-specific settings with the current gene settings.

Option	Steps

To immediately use the BED file as a panel	1. Click Import as Panel.
	
	  The Panel Name dialog box opens.
	
	Figure 11-4: Panel Name dialog box
	
	2. Enter the name for the panel, and then click OK.
	
	  The Panel Name dialog box closes, and the Manage Panels dialog box opens. The full directory path to the imported panel, including the panel name, is displayed in the Import from Panel File field.
	
	3. Continue to "Importing a Panel" on page 105.

8. Click OK.

   The Save As dialog box opens. By default, the BED file is named default.bed.

9. Edit the name of the BED file as needed, and save it in a location of your choosing.

Convert VCF File to a BED File Tool

If you do not have a needed BED file for a panel, you can use the Convert VCF to BED tool to convert a sample VCF file to the required BED file. The BED file contains the appropriate regions for the variants that are found in the VCF file.

To convert a VCF file to a BED file

1. On the Geneticist Assistant main menu, click Tools > Convert VCF File to BED File.

   The Convert VCF to BED dialog box opens.

Figure 11-5: Convert VCF to BED dialog box
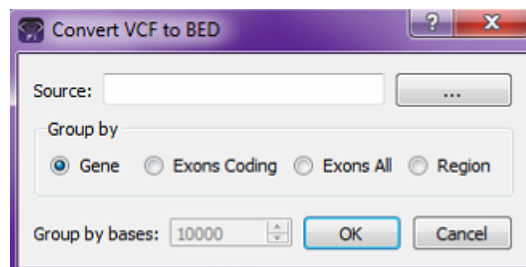
2. Next to the Source field, click the Browse button […] to open the Open dialog box, and then browse to and select the appropriate VCF file.

3. Indicate how the regions are to created in the BED file.

Option	Description
Gene	Adds the entire region for the gene in which each variant is found to the BED file. For example, if the variant is found in the BRCA1 gene, then the entire BRCA1 region is added to the BED file.
Exons Coding	Adds the coding exon for each variant in the VCF file as a region in the BED file.
Exons All	Adds both the coding exons and the non-coding exons for each variant in the VCF file as a region in the BED file.
Region	Creates a region for each variant found in the VCF file, based on the variant's position. If you select Region, then you must also specify the total number of bps that are to be found in each region (the Group by Bases value) with one half of the total number of bps found on each side of the variant position. For example, if you specify 10,000 for Group by Bases, then 5000 bps are found on either side of the variant position for the BED file region.

4. Click OK.

   The Save as dialog box opens.

5. In the Save as dialog box, enter a name and select a location in which to save the BED file, and then click Save.

The default name for the BED file is default.BED and the default location in which to save the BED file is the same directory as the source VCF file, but you can change one or both of these values. The file type is .bed and you cannot change this.

A Convert VCF to BED file message opens, indicating that the conversion is completed.

Figure 11-6:   Convert VCF to BED file message
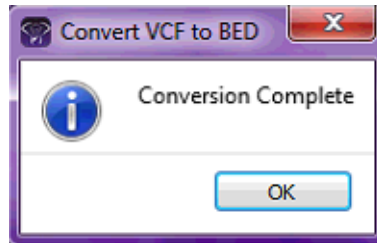
6. Click OK.

The message closes. The Convert VCF to BED dialog box remains open.

7. Click "x" to close the Convert VCF to BED tool.

Deleting 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 11-7:   Delete Pathogenicity Changes dialog box
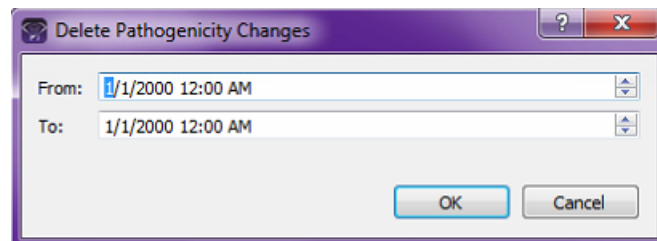
2. Change the From value, the To value, or both values as needed. To change the value, select the value that you are changing, for example, the month, and then you can manually enter the new value, or you can use the Up/Down field arrows to change the value.

   You must select each value that are you changing one at a time. If you need to confirm the time period, remember that the Pathogenicity Changes pane on the Variant tab displays the date and time that a pathogenicity change was made. See "Pathogenicity Changes pane" on page 251.

3. Click OK.

A Confirm Deleting Pathogenicity Changes message opens, asking you if you are sure that you want to delete "x" number of pathogenicity changes within the date range.

Figure 11-8:   Confirm Deleting Pathogenicity Changes message
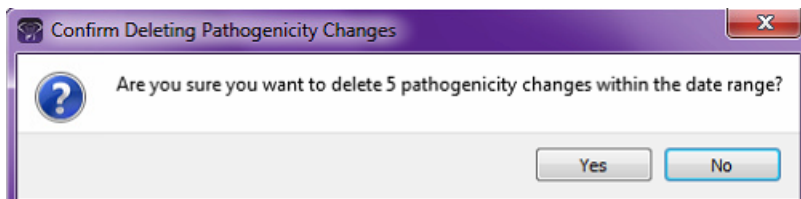
4. Click Yes.

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, variants comments and/or variant artifact types, 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 and/or set the artifact types. For either situation, this means that if multiple variants have the same pathogenicity and/or artifact type for a panel group, you do not have to set the pathogenicity of each variant individually. Instead, you can use this tool to set the same pathogenicity and/or artifact type for all the variants at once.

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 11-9:   Import Variants dialog box
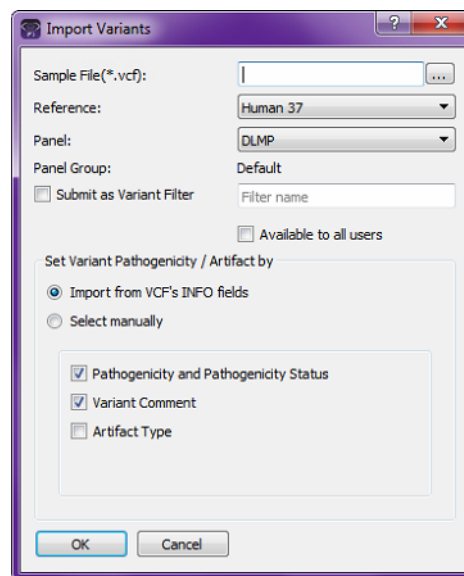
2. Next to the Sample File field, click the Browse button [...] to open the Select a Variant File dialog box, and then browse to and select the appropriate VCF file.

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' pathogenicities, comments, and/or artifact types.

   The Panel group name is displayed for Panel Group.

5. Optionally, to create a filter based on the variants in the VCF file, do the following:

   a. Select Submit as Variant Filter, and then in the Filter name field, enter a name for the filter.

   b. To make the filter available only to you, then leave Available to all users cleared; otherwise, to make the filter available to any and all users, select this option.

---

*Remember, you apply this variant filter through the Load Filters option. See *

6. Continue to one of the following:

   • below.

   •

To import the variants' pathogenicities, pathogenicity statuses, comments, and/or artifact types

1. Leave Import from VCF's INFO fields selected.

2. Do either or both of the following:

   • 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.

   • If the VCF file contains the artifact types for the variants, then to import this information, select this option; otherwise, if the VCF file does not contain this information, or you do not want import this information, then leave this option cleared.

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 define the variants' pathogenicities, pathogenicity statuses, comments, and/or artifact types

When you manually define the pathogenicities, statuses, and/or artifact types for the variants, these properties 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 and/or setting the artifact type for the variants.

Figure 11-10: Import Variants dialog box, Select manually option
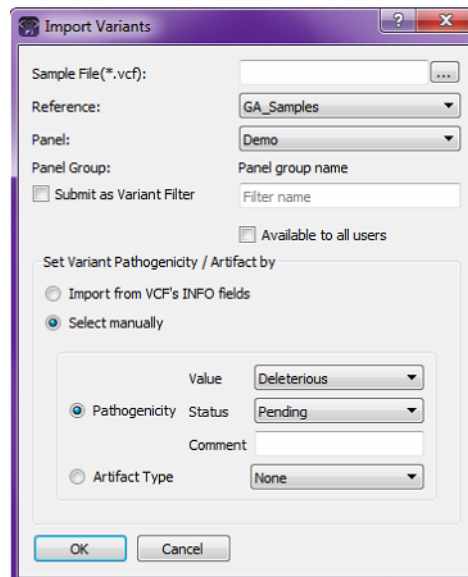
2. Do any or all of the following:

   • 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 in the Geneticist Assistant Database" on page 55.

   • In the Pathogenicity Comment field, enter a common comment for all the variants.

   • Select Artifact Type, and then on the Artifact Type dropdown list, select a common artifact type 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 are now displayed with the specified information (such as the pathogenicity) in any samples that contain these variants.

Find Samples with Variants Tool

You can use the Find Samples with Variants tool to find all the samples in your Geneticist Assistant database that contain specific variants. You can search for the variants based on the Variant database ID, the variant rs identification from dbSNP, or the variant description.

To find samples with specific variants with the Find Samples with Variants tool

1. On the Geneticist Assistant main menu, click Tools > Find Samples with Variants.

   The Find Samples with Variants dialog box opens.

   Figure 11-11: Import Variants dialog box
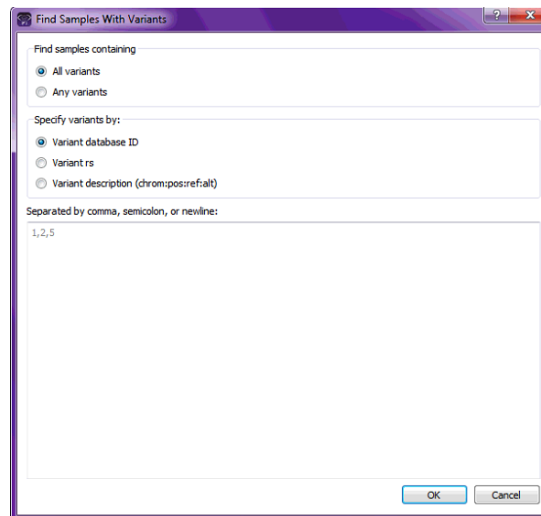
2. Indicate which samples that the database search is to return:

   • All variants - Selected by default. Return only those samples in which all the indicated variants were found.

   • Any variants - Return any sample in which at least one of the indicated variants was found.

3. Indicate how to search for the variants based on one of the following:

   • The Variant database ID, which is displayed in the ID column in the various Variant tables in Geneticist Assistant.

   • The variant rs identification from dbSNP, which is displayed in the rs column of the various Variant tables in Geneticist Assistant.

   • The variant description, which includes the following four fields separated by colons: chromosome, chromosome position, reference allele, and alt allele.

4. Enter the variant search criteria.

When searching for multiple variants, you must separate the variant search criteria by a comma or a semi-colon, or place each search criterion on its own line.

5. Click OK.

A list of all the samples that contain the specified variants is displayed on a Samples with Variants tab.

Figure 11-12: Samples with Variants tab

The context menu for this tab is identical to the context menu for the Samples pane on the Run tab. See "The Samples Pane (Run tab) Context Menu" on page 191.

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 11-13: Merge BAM Files dialog box
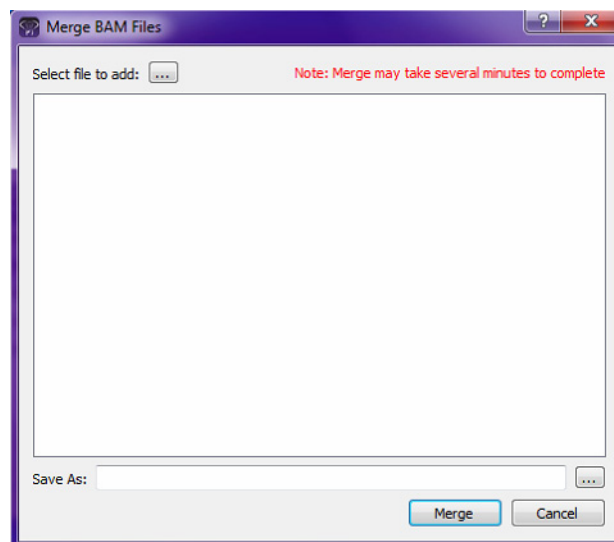
2. At the top of the dialog box, click the Browse button [...] to open the Select a file to add dialog box, and browse to and select the BAM files that are to be merged.

   CTRL-click 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 12

The Geneticist Assistant Customer Portal and Submissions

The Geneticist Assistant customer portal is a web-based platform that serves as a dedicated hub for tracking and managing your organization's patient and sample information and the tests that have been carried out for each patient/sample combination (referred to as a submission). You designate specific users, known as *web users*, to have login and read/write permissions to this platform. These web users can enter your organization's patient and sample information without ever having to log in to an instance of Geneticist Assistant. The patient and sample information is directly linked between the Geneticist Assistant database and the customer portal. Any patient that is in the Geneticist Assistant database is automatically available for selection in the portal, and if a new patient must be created to submit information, then this patient is automatically added to the Geneticist Assistant database. A barcode is generated for each submitted patient/sample combination, and you can use this barcode to validate that the correct patient and sample combination was submitted to the customer portal and to confirm sample identity when you are reviewing a sample in Geneticist Assistant. A full record of every submission that any user has ever made through your organization's secure access point to the Geneticist Assistant customer portal is displayed on the Submissions tab.

This chapter covers the following topics:

- "Logging in to the Geneticist Assistant Customer Portal" on page 307.

- "The Geneticist Assistant Customer Portal Layout" on page 310.

- "Submitting Patient/Sample Tracking Information through the Geneticist Assistant Customer Portal" on page 312.

- "The Submissions Tab" on page 315.

Logging in to the Geneticist Assistant Customer Portal

To log in and use the customer portal, two files are required:

- Cert.pem file, which you must save in C:\Program Files\SoftGenetics\SoftGenetics Server\ga on the Geneticist Assistant server.

- Cert.pfx, which you can save anywhere on the Geneticist Assistant client. You must double-click this file to launch the Certificate Import Wizard and import the encryption key for your Geneticist Assistant installation.

If your organization already uses encryption for patient data, then these files should already be configured and available for use; otherwise, work with your organization's System Administrator to create and configure these files, or you can contact.tech_support@softgenetics.com for assistance.

By default, your Geneticist Assistant Administrator account (the Administrator role that you were prompted to create for the installation of Geneticist Assistant) is designated as a user of the customer portal. The username and password for logging in to the customer portal are identical to the username and password that were specified for the Administrator role. All other users of the customer portal must be specified as *web users*. (See "Managing Web Users" on page 98.)

Figure 12-1:   Geneticist Assistant Login dialog box with credentials for the Geneticist Assistant customer portal
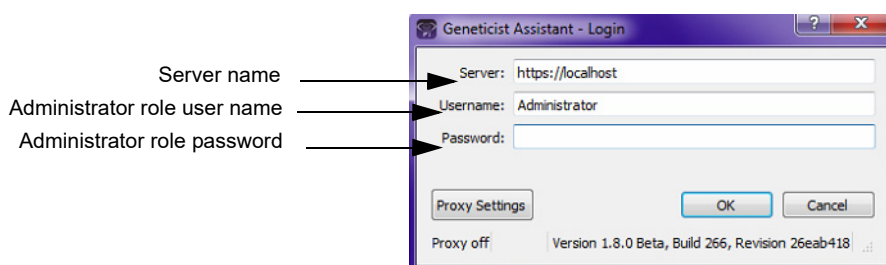
Server name

Administrator role user name

Administrator role password

The login URL for the Geneticist Assistant customer portal is set to the following:

https://<server name>/samplesub

where <server_name> is the name of your Geneticist Assistant server, for example, localhost.

To log in to the Geneticist Assistant customer portal

1. Open a browser of your choice.

2. In the URL field, enter the login URL for your Geneticist Assistant customer portal.

The login URL for the Geneticist Assistant customer portal is set to the following: https://<server name>/samplesub, where <server_name> is the name of your Geneticist Assistant server, for example, localhost.

The Customer Portal Login page opens.

Figure 12-2:   Customer Portal Login page
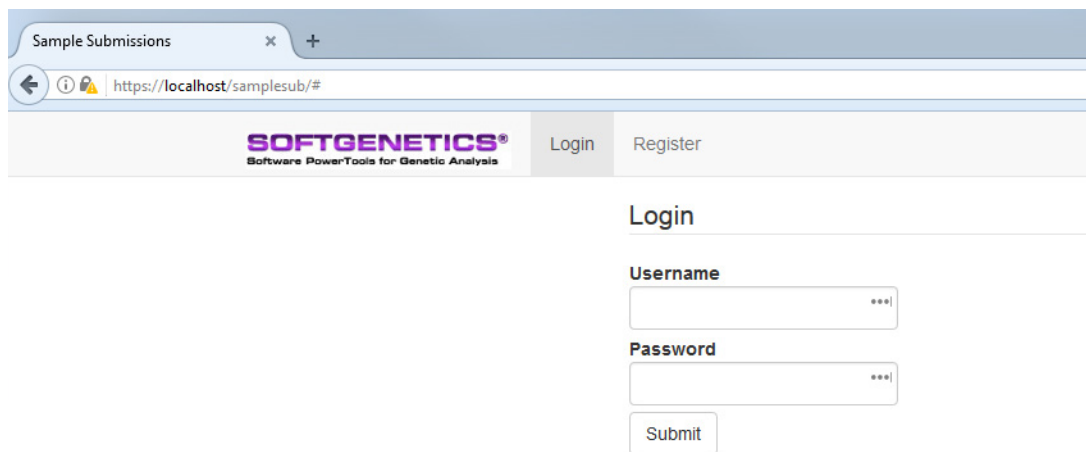
3. Enter your username and password, and then click Submit.

By default, your Geneticist Assistant Administrator account (the Administrator role that you were prompted to create for the installation of Geneticist Assistant) is designated as a user of the customer portal. The username and password for logging in to the customer portal are identical to the username and password that were specified for the Administrator role. If you are not the Administrator, then you must be specified as a web user. (See "Managing Web Users" on page 98.)

The Customer Portal opens. The Patients and Samples page is the open page. All the patients that are currently saved in your Geneticist Assistant database are listed in the Patients pane on the left side of the page. Any patient/sample combination that has been submitted to the customer portal is displayed in the Samples pane on the right side of the page. See Figure 12-3 on page 309.

Figure 12-3:   Geneticist Assistant Customer portal, Patients and Samples page
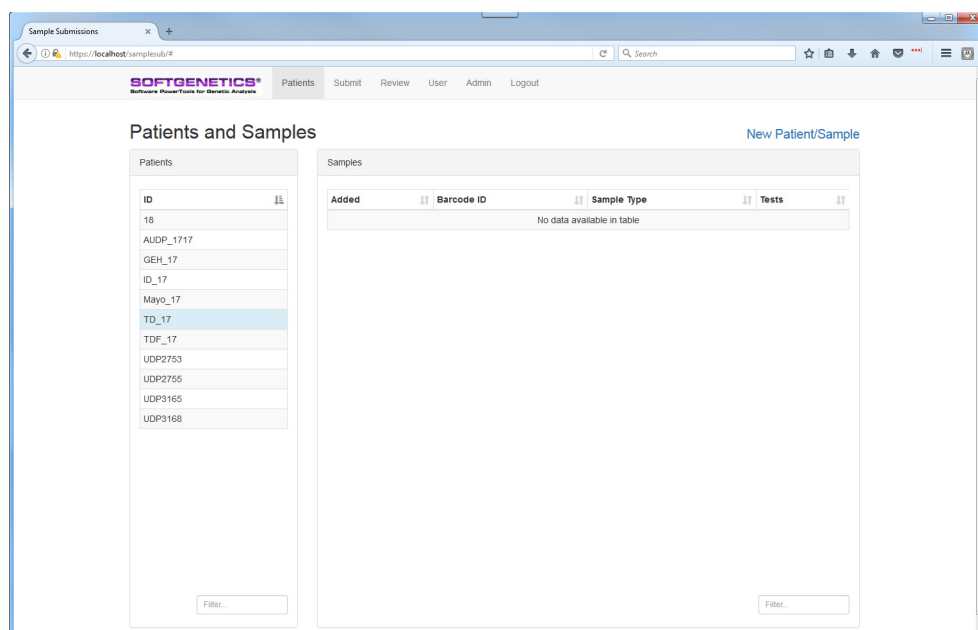
4.   Continue to "The Geneticist Assistant Customer Portal Layout" on page 310.

The Geneticist Assistant Customer Portal Layout

Figure 12-4:   Geneticist Assistant customer portal, Patients and Samples page
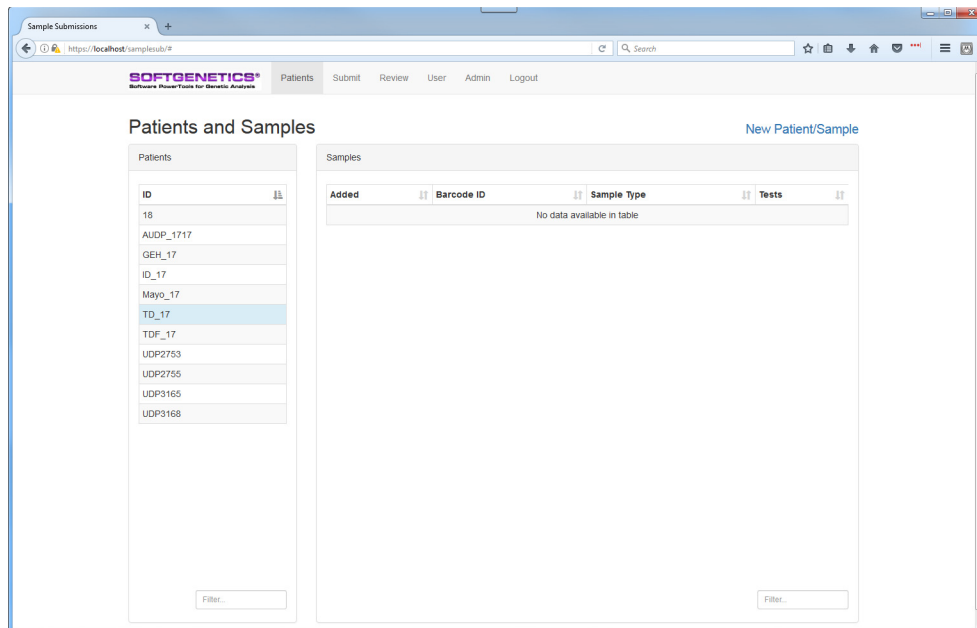
The customer portal main menu is displayed at the top of every customer portal page. The main menu has the following options:

Main Menu Option	Description
Patients	Opens the Patients and Samples page. This is the page that opens after you log in to the Geneticist Assistant customer portal. All the patients that are currently saved in your Geneticist Assistant database are listed in the Patients pane on the left side of the page. Any patient/sample combination that has been submitted to the customer portal for tracking purposes is displayed in the Samples pane on the right side of the page.
Submit	Opens the Submit Sample page. This is the page on which you enter the information for the patient and sample combination that you are submitting to the Geneticist Assistant customer portal. This page has seven distinct sections for entering the information for the patient and sample combination that you are submitting to the Geneticist Assistant customer portal. Your organization's Administrator is responsible for customizing the fields that are displayed in these sections, including whether a field is enabled or disabled, and whether a field is required or optional. See "Admin" on page 311.
Review	Opens the Review page, where you can review a patient and sample combination and verity that the information that has been entered for it is correct before you submit the information to the customer portal.

Main Menu Option	Description
User	Opens the User page. You use this page to enter the contact details for the providers and institutions that are to be available for inclusion with any submission including the ordering provider, the genetic counselor, the referring hospital or laboratory, and the institutional billing. Your organization's Administrator is responsible for customizing the fields that are displayed in these sections, including whether a field is enabled or disabled, and whether a field is required or optional. See "Admin" below.
Admin	Available only to the default Administrator user for your Geneticist Assistant installation. Opens the Admin page where the Administrator of your organization's customer portal specifies the fields that are to be enabled/disabled on the Submit and User pages, and whether an enabled field is to be required or optional. In addition to specifying the field rules for the Submit and User pages, an Admin can also customize the barcode that is automatically generated for a submission. This barcode can be used to validate that the correct patient and sample information combination was submitted to the customer portal and to confirm sample identity when you are reviewing a sample in Geneticist Assistant. "Submitting Patient/Sample Tracking Information through the Geneticist Assistant Customer Portal" on page 312. **Note:** If you need assistance with customizing your Geneticist Assistant customer portal interface, contact tech_support@softgenetics.

Submitting Patient/Sample Tracking Information through the Geneticist Assistant Customer Portal

You must be the Administrator of Geneticist Assistant, or designated as a web user to log in and submit information through Geneticist Assistant customer portal. A full record of every submission that any user has ever made through organization's secure access point to the Geneticist Assistant customer portal is displayed on the Submissions tab.

To submit patient/sample tracking information through the customer portal

1. Log in to the customer portal.

   By default, your Geneticist Assistant Administrator account (the Administrator role that you were prompted to create for the installation of Geneticist Assistant) is designated as a user of the customer portal. The username and password for logging in to the customer portal are identical to the username and password that were specified for the Administrator role. If you are not the Administrator, then you must be specified as a web user. (See "Managing Web Users" on page 98.)

2. On the Patients and Samples page, click New Patient/Sample, or on the main menu, click Submit.

   The Submit Sample page opens. The Patient and Specimen section is displayed. The Sections pane on the left side of the page lists the seven sections for the page by name in which you can enter the patient/sample tracking information.

Figure 12-5:   Geneticist Assistant customer portal, Submit Sample page, Patient and Specimen section

Your organization's Administrator is responsible for customizing the fields that are displayed in all the sections on the Submit Sample page, including whether a field is enabled or disabled, and whether a field is required or optional.

3. In the Patient and Specimen section, on the Saved Patients dropdown list, do one of the following:

   • Leave the list set to Add New, and then enter the new patient information.

   • Select a patient from your organization's Geneticist Assistant database.

The patient and sample information is directly linked between the Geneticist Assistant database and the customer portal. Any patient that is in the Geneticist Assistant database is automatically available for selection on the Saved Patients dropdown list. If you add a new patient, then this new patient is automatically added to your organization's Geneticist Assistant database and going forward, is available for selection in the portal.

4. Continue opening each section on the page, and then enter the appropriate information for the patient/sample combination.

5. Optionally, after you have entered all the patient/sample tracking information, in the Sections pane on the Submit Samples page, click Review to open the Review Submission page and review the information and make any necessary changes before submitting it.

6. On the Submit Samples page, in the Sections pane, click Submit.

   The patient/sample information is submitted to the customer portal. The submission is automatically recorded on the Submissions tab in Geneticist Assistant. The Submit Sample page remains open. A Submission Successful message is displayed on the page along with two options: Download Packing Slip and Download Submission Details.

Figure 12-6:   Submission Successful message with options
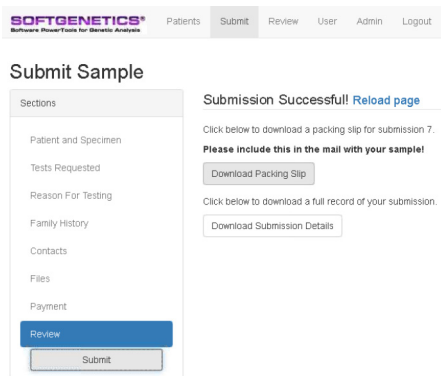
7. Optionally, click either one or both options to download the needed information for the submission:

The prompt that opens and the location in which the file is automatically saved is dependent on the browser that you are using to access the customer portal.

- Download a packing slip: Generates a packing slip in PDF format that includes a barcode.

  If your organization uses barcodes, then you can scan this barcode and validate that the correct patient and sample combination was submitted to the customer portal and to confirm sample identity when you are reviewing a sample in Geneticist Assistant.

Figure 12-7:   Packing slip with barcode for patient/sample information submitted to the Geneticist Assistant customer portal

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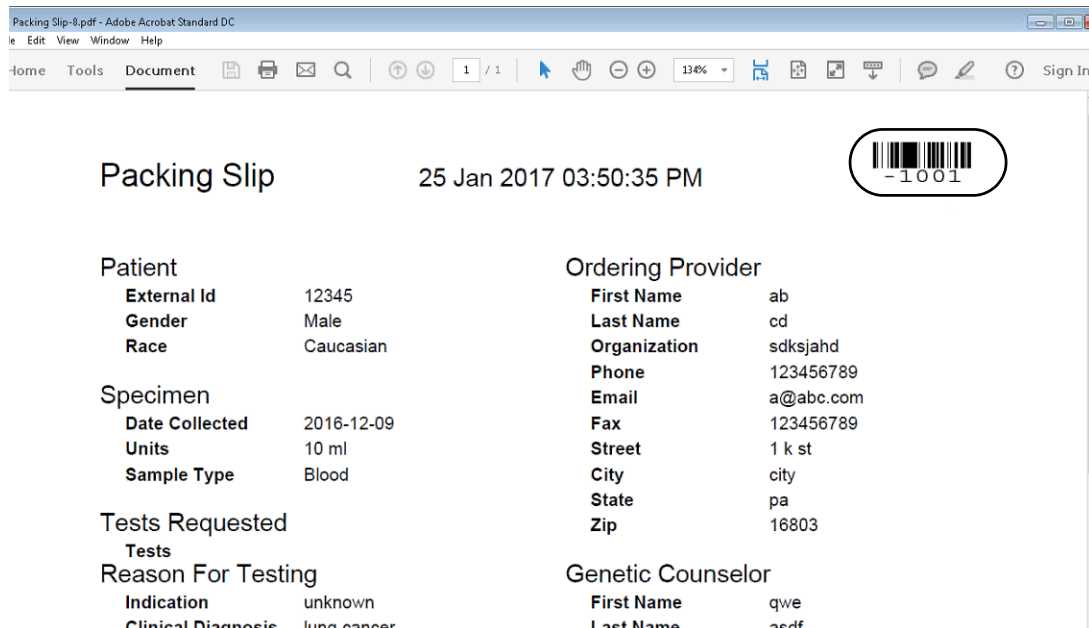
You can always choose to generate a packing slip with a barcode for this or any other submission at a later date. To do so, on the customer portal main menu, click Review to open the Review Sample page, select the appropriate patient/sample combination, and then click Download Submission Packing Slip.

- Download Submission Details: Generates a full record of the submission in PDF format. The information that is displayed on the Submissions Detail form is identical to the information that is displayed on the Packing Slip form, but without the barcode.

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*You can always view the full record of any submission on the Submissions tab in Geneticist Assistant. See *

The Submissions Tab

A full record of every submission that any user has ever made through your organization's secure access point to the Geneticist Assistant customer portal is displayed on the Submissions tab in Geneticist Assistant. To open the Submissions tab, on the Geneticist Assistant main menu, click Tabs > Submissions.

Figure 12-8:   Submissions tab

To open and view all the details for an individual submission, double-click the submission on the Submissions tab. Each pane on the Submission tab corresponds to specific information that was entered for the submission, for example, Patient, Specimen, and so on. The information always includes the barcode for the submission, which you can use to comfirm sample identity.

Figure 12-9:   Submission tab

To use the barcode to confirm sample identity

If a packing slip has been generated for a submission, then two options are available for using the barcode to validate that the correct patient/sample combination was submitted to the customer portal and to confirm sample identity when you are reviewing a sample in Geneticist Assistant:

• On the Submission tab, click Verify Barcode, and then use a barcode scanner to scan the barcode on the physical packing slip.

• On the Submission tab, click Verify Barcode, and then enter the barcode number that is displayed on the packing slip in the field. If the barcode number that you enter matches the barcode number for the submission, then "Match" is displayed in green above the Verify Barcode number field.

Figure 12-10: Verifying submissions using a manually entered barcode number

Appendix A
Common Procedures

Regardless of the entity with which you are working in Geneticist Assistant (a run, a patient, a sample, or a variant), there are several procedures common to all these entities. This appendix describes these common procedures and where appropriate, details the slight differences that can exist based on the entity for which you are carrying out the procedure.

This appendix covers the following topics:

- "Reviewing Panel Statistics" on page 319.

- "Setting Read and Write Permissions" on page 326.

- "Managing a Batch Variants Report" on page 329.

- "Generating a Batch Coverage Report" on page 335.

- "Creating an Action List" on page 338.

- "Comparing Samples in a Run" on page 340.

- "Adding a Variant Interpretation" on page 342.

Appendix A
Common Procedures

Reviewing Panel Statistics

Panel statistics are displayed on the Panel Statistics tab. The information that is displayed on the Panel Statistics tab is the same (using all samples by default) *except* if you have selected multiple samples from the Run tab or multiple associated samples on the Variant tab. You can open and review the information on the Panel Statistics tab from the following locations in Geneticist Assistant:

• The Panel Statistics tab from the Panels menu.

• A patient sample when viewed from the Samples sub-tab on the Patient tab.

• A panel that was used for a run when viewed from the Runs tab.

• A panel that was used for a run when viewed from Samples pane on the Run tab.

• A sample's run when viewed from the Associated Panels pane on the Variant tab.

To review panel statistics

When reviewing the information on the Panel Statistics tab, you can filter the information by the following criteria:

• All panel samples, or selected panel samples.

• Samples that have been added to the panel within the last 30 days of the current day's date, or a custom date range.

• Selected runs for the panel.

When the Panel Statistics tab first opens, the Coverage Regions sub-tab is the open tab. You can select a different panel from the Panel dropdown list to view statistics for a different panel. You can also select a different panel to change the panel group, but you cannot change the panel group directly.

Figure A-1:   Coverage Regions sub-tab (Panel Statistics tab)

1. To display data on the Panel Statistics tab, do one of the following:

To display Panel Statistics information from:	Steps
The Panel Statistic tab from the Panels menu	1. On the Geneticist Assistant main menu, click Panels > Panel Statistics. The Panel Statistics tab opens. 2. On the Panels dropdown list, select the panel for which the statistics are to be viewed.
The Samples sub-tab on the Patient tab	In the Samples pane, select a sample or CTRL-click to select multiple samples, and then right-click on any selected sample and on the context menu that opens, click Panel Statistics.
The Runs tab	1. Select a run, or CTRL-click to select multiple runs. 2. Right-click any selected run, and on the context menu that opens, click Panel Statistics.
The Run tab	In the Samples pane, select a sample or CTRL-click to select multiple samples, and then right-click on any selected sample and on the context menu that opens, click Panel Statistics. **Note:** If you select a small number of samples, such as two to four, then, by default, the panel statistics are calculated based only on the selected samples and before the statistics are calculated, a Very few samples message dialog box opens, warning you of this and asking you if you want to continue. You can click Yes, or you can click Cancel to return to the Run tab and select more samples.
The Associated Samples pane on the Variant tab	1. Select a sample, or CTRL-click to select multiple samples. 2. Right-click any selected sample, and on the context menu that opens, click Panel Statistics. **Note:** If you select multiple samples from multiple runs for different panels, then a Select Panel dialog box opens. You must first select the appropriate panel before the statistics can be calculated. **Note:** If you select a small number of samples, such as two to four, then, by default, the panel statistics are calculated based only on the selected samples and before the statistics are calculated, a Very few samples message dialog box opens, warning you of this and asking you if you want to continue. You can click Yes, or you can click Cancel to return to the Variant tab and select more samples.

2. Optionally, edit any or all of the calculation criteria as needed, and then click OK to recalculate the panel statistics and update the display accordingly.

Option	Description
All panel samples	If you select this option, then all samples that have been added to the panel are displayed. To specify a custom date range, select added between, and then change one or both dates as needed. To change the dates, you can do either of the following: • You can select any part of the date that you want to change, or the entire date, and then manually enter the new value. (The default date range is the last 30 days). • You can select any part of the date that you want to change, and then use the Up/Down arrows to change the date accordingly.
Selected runs	To select specific runs for the panel, choose Selected runs, and then next to Selected runs, click the Browse button ⟨...⟩ that to open the Select Runs dialog box. All the runs for the selected panel are displayed in the dialog box. Select a run, or CTRL-click to select multiple runs, and then click OK. *Figure A-2:* *Select Runs dialog box*

Option	Description
Selected samples	To select only specific samples for the panel, choose Selected samples, and then next to Selected samples, click the Browse button `...` that to open the Select Samples dialog box. All the samples for the selected panel are displayed in the dialog box. Select a sample, or CTRL-click to select multiple samples, and then click OK. *Figure A-3:*　　*Select Samples dialog box*

3. Optionally, after calculating the panel statistics, continue to any of the following needed to view information on any of the five sub-tabs that are available for the Panel Statistics tab:

- Coverage Regions sub-tab. See "Coverage Regions sub-tab (Panel Statistics tab)" on page 323.

- Samples sub-tab. See "Samples sub-tab (Panel Statistics tab)" on page 323.

- Variants sub-tab. See "Variants sub-tab (Panel Statistics tab)" on page 324.

- Patients sub-tab. See "Patients sub-tab (Panel Statistics tab)" on page 324.

- Reports sub-tab. See "Reports sub-tab (Panel Statistics tab)" on page 325.

Coverage Regions sub-tab (Panel Statistics tab)

The Coverage Regions sub-tab has a single pane, the Coverage Regions pane, that lists the coverage information across all the samples, for the selected run or runs, or for the selected sample or samples for every region in the selected panel and panel group. See "Coverage Regions tab" on page 200 for a detailed discussion of the information that is displayed on this sub-tab.

Figure A-4:   Coverage Regions sub-tab (Panel Statistics tab)

Samples sub-tab (Panel Statistics tab)

The Samples sub-tab has a single pane, the Samples pane, that lists all the samples that were imported for the panel or in the selected run or runs or for the selected sample or samples for the selected panel and panel group. You can double-click a sample on this sub-tab to open the Sample tab. (See Chapter 8, "Geneticist Assistant Data Review - Sample Tab," on page 209.) 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 "The Samples Pane (Run tab) Context Menu" on page 191.

Figure A-5:   Panel Statistics tab, Samples sub-tab

Variants sub-tab (Panel Statistics tab)

The Variants sub-tab has a single pane, the Variants pane, that lists all the variants that were detected for the panel in all the samples that were imported in the selected run or runs or for the selected sample or samples for the selected panel and 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. (See Chapter 9, "Geneticist Assistant Data Review - Variant Tab," on page 243.) 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 variant on the Sample Variant Data sub-tab. (See "Sample Variant Data Sub-Tab" on page 212.)

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

Patients sub-tab (Panel Statistics tab)

The Patients sub-tab has a single pane, the Patients pane, that lists all the patients who were associated with all the samples the panel or the selected sample or samples or for the selected run or runs that were imported for the selected panel and panel group. You can double-click a patient in the pane to open the corresponding Patient tab. You can also right-click 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 137.

Figure A-7:   Panel Statistics tab, Patients sub-tab

Reports sub-tab (Panel Statistics tab)

The Reports sub-tab contains a single pane, the Reports pane, that lists all the reports that were generated for the panel or for the selected run or runs or selected sample or samples 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 includes these same reports.

Figure A-8:   Panel Statistics tab, Reports sub-tab
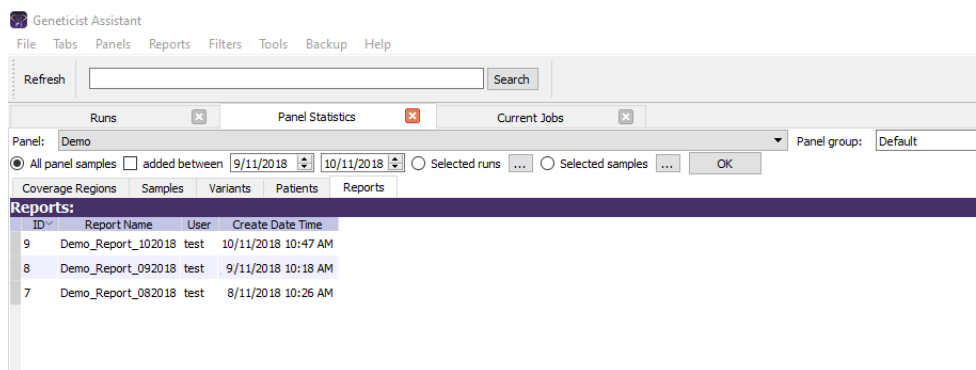
You can right-click anywhere on this sub-tab to open a context menu with options that are identical to the options that are on context menu for the Reports pane on the Patient sub-tab on the Patient tab. See "Patient tab, Patient sub-tab, Reports pane" on page 154.

For detailed information about generating and saving Panel Statistics reports, see Chapter 10, "Geneticist Assistant Reports," on page 265.

Setting Read and Write Permissions

Permissions determine the Read and/or Write functions that are available to selected users or groups for a patient, sample, or run. You can set Read/Write permissions for these Geneticist Assistant entities from the following locations:

- Patient: The Patients tab and the Patient sub-tab on the Patient tab.

- Patient Sample - The Sample sub-tab on the Patient tab.

- Run - The Runs tab.

- Run Sample - The Samples pane on the Run tab (per sample basis).

- Sample in which a selected variant was found - Associated Samples pane on the Variant tab.

You can set permissions for any entity if you are an Administrator user. If you are a standard user, then set the permissions for an entity that you added to the Geneticist Assistant database or to which you have been granted read/write permissions.

To set Read and Write permissions for a patient, sample, or run

1. Open the location from which you are setting the permissions:

   - The Patients tab.

   - The Samples sub-tab on the Patient tab.

   - The Runs tab.

   - The Samples pane on the Run tab.

   - The Associated Samples pane on the Variant tab.

2. Do one of the following:

   - Select the patient or CTRL-click to select the multiple patients for which you are setting the permissions, and then right-click any selected patient, and on the context menu that opens, click Permissions.

   - Select the sample or CTRL-click to select the multiple samples for which you are setting the permissions, and then right-click any selected sample, and on the context menu that opens, click Permissions.

   - Select the run or CTRL-click to select the multiple runs for which you are setting the permissions, and then right-click any selected run, and on the context menu that opens, click Permissions.

   The Permissions dialog box opens. The first field on the dialog box indicates the entity (Patient, Sample, or Run) for which you are setting the permissions. The dialog box also displays the names of all the users and/or groups and their assigned permissions for the patient, sample, or run. If you opened the Permissions dialog box from the Runs tab, then

an additional option, Apply permission settings to samples, is displayed.

Figure A-9:   Permissions dialog box (Patient or Sample)
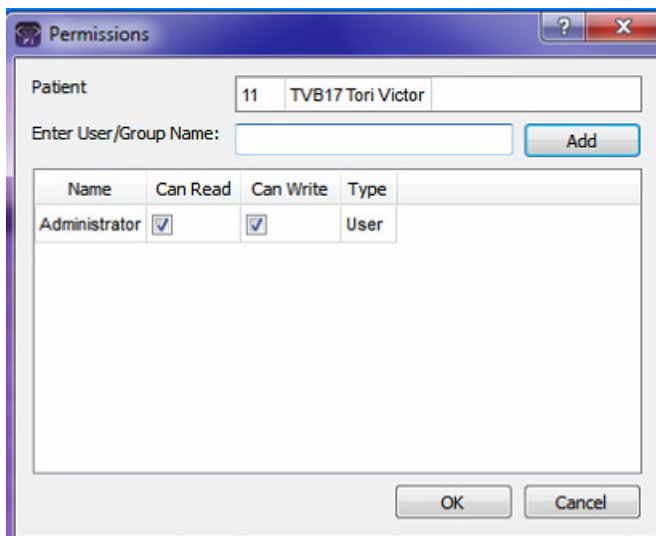
Figure A-10:   Permissions dialog box (Runs tab)
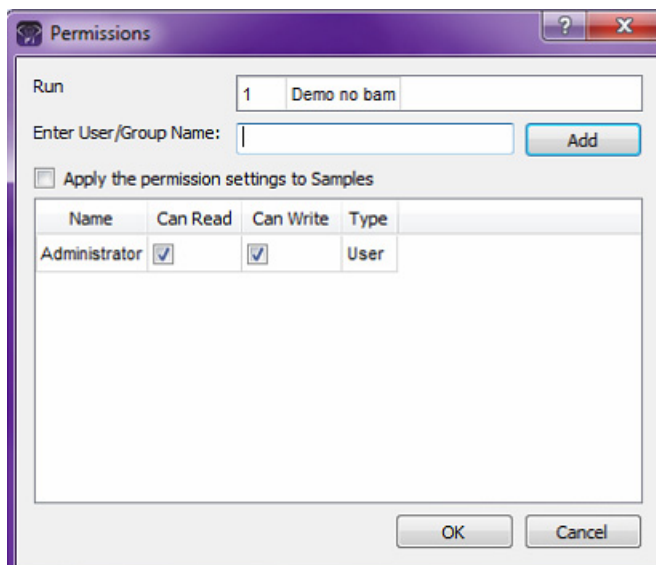
3. Edit the permissions for any user or group as needed and then click OK; otherwise, to
    add permissions for a new user or group, go to

---

*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. Note the following about the search string:

   • The search string is not case-sensitive, but the string must begin with the user or group name. For example, a search string of "Ja" or "ja" returns a user with the name of Jane Doe, but a search string of "Doe" does not.

   • 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 a user name or group name from the list, and then click Add.

   The user name or group name is displayed in the lower pane of the dialog box. The Can 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 records, samples, or runs. They cannot edit them.

6. Optionally, if the user or group must be able to edit the patient records, samples, or runs as well, then select the Can Write permission.

7. If you are setting the permissions for one or more runs, then optionally, if all users and groups are to have their same run permissions applied to *all* the *samples* in all the selected runs, then select Apply the permission settings to Samples; otherwise, leave this option cleared, or clear it if necessary.

8. Click OK.

   A Permission Updated message opens.

9. Click OK.

   Both the message and the Permissions dialog box close. The specified permissions are set for the patient records, samples, or runs.

Managing a Batch Variants Report

A *Batch Variants report* is a report that includes information for variants in a selected sample or samples. You can create multiple reports for multiple samples in a single step. A *Batch Variants report configuration* determines the data that you are including in the report. You choose if the configuration is to be available only to the user who created it, or to all users. The configuration does not include the report format 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 Batch Variants report.

Figure A-11:   Batch Variants Report dialog box
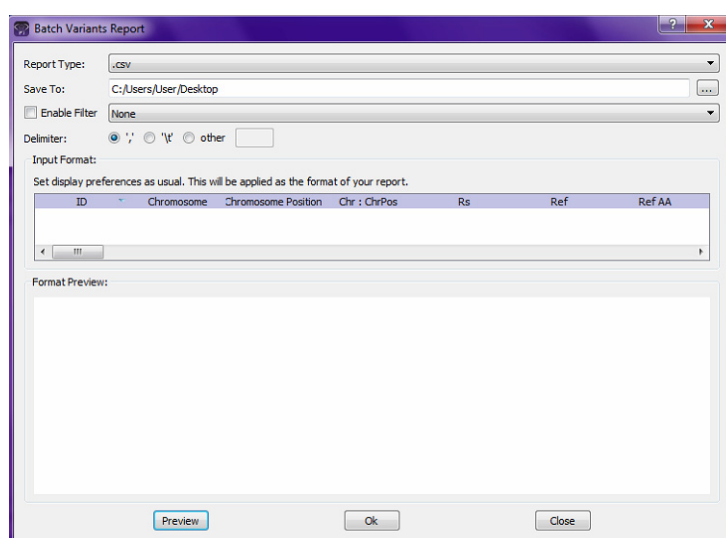
You can generate a Batch Variants report for a selected sample or samples from the following locations in Geneticist Assistant:

• The Samples sub-tab on the Patient tab.

• The Runs tab. A separate report is saved for each sample of the selected run.

• The Samples pane on the Run tab.

• The Associated Samples pane on the Variant tab.

Managing a Batch Variants report includes generating the report according to a new or saved configuration, editing a report configuration, or deleting a report configuration. See:

To generate a Batch Variants report based on a new configuration

The name for a Batch Variants report is automatically assigned based on the sample ID number and sample name for each sample for which you are generating the report (for example, 76_800458.variants.filter) and you cannot change this. You must select a location in which to save the report. A separate report is saved for each sample selected.

1. Open the location from which you are generating the report:

   • The Samples sub-tab on the Patient tab.

   • The Runs tab.

   • The Samples pane on the Run tab.

   • The Associated Samples pane on the Variant tab.

2. Do one of the following:

   • Select the sample or CTRL-click to select the multiple samples for which the Batch Variants report is to be generated, and then right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.

   • Select the run or CTRL-click to select the multiple runs for which the Batch Variants report is to be generated, and then right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants report.

   The Batch Variants report dialog box opens. See Figure A-11 on page 329.

3. On the Report Type dropdown list, select the report type. (The default value is .csv)

4. 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, the default value of C:/Users/<User>/Desktop is displayed. Do one of the following:

   • Leave this location as-is, and then go to Step 5.

   • 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.

5. Optionally, to filter the data that is to be displayed in the report, select Enable Filter, and then from the Filter dropdown list, select the appropriate pre-configured filter.

For detailed information about filtering the data that is displayed in a report, see "To filter the data that is displayed in a tab pane" on page 32.

6. By default, "comma" is selected for the report delimiter. You can leave this value as-is, or you can select "tab," or "other." If you select "other," then a blank field is enabled and you must specify the delimiter, for example, an underscore (_) in this field.

7. Optionally, to set the display preferences for the report, do any of the following:

   • Rearrange the data columns.

   • Rename a column header.

   • Show/hide columns (manually or by loading a configuration).

   For detailed information about working with data columns, including moving and renaming columns, or showing/hiding columns, see "Setting Display Preferences in the Geneticist Assistant Main Window" on page 31.

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

9. Click OK.

   The Batch Variants report is generated for all selected samples. After the report is successfully generated, a Batch Report Finished message opens, indicating that the report was generated and where the report was saved.

Figure A-12:   Batch Report Finished message
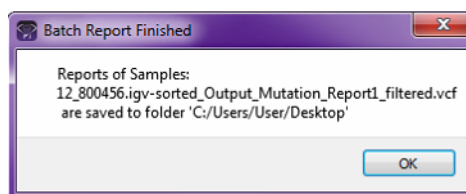
10. Click OK.

    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. Open the location from which you are generating the report:

   • The Samples sub-tab on the Patient tab

   • The Runs tab.

   • The Samples pane on the Run tab.

   • The Associated Samples pane on the Variant tab.

2. Do one of the following:

   • Select the sample or CTRL-click to select the multiple samples for which the Batch Variants report is to be generated, and then right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.

- Select the run or CTRL-click to select the multiple runs for which the Batch Variants report is to be generated, and then right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants report.

The Batch Variants report dialog box opens. See Figure A-11 on page 329.

3. On the Batch Variants Report dialog box, select the output format for the report.

4. 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, the default value of C:/Users/<User>/Desktop is displayed. Do one of the following:

- Leave this location as-is, and then go to Step 5.

- 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.

5. Right-click on any heading in the Input Format pane, and then choose Select Columns.

The Select Columns dialog box opens.

6. 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 A-13:   Load Configuration dialog box
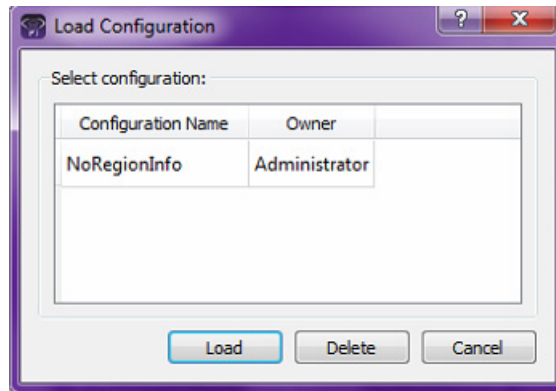
7. Select a configuration, and then click Load.

The Load Configuration dialog box and the Select Columns dialog box close.

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 for all selected samples. After the report is successfully generated, a Batch Report Finished message opens, indicating that the

report was generated and where the report was saved.

Figure A-14:   Batch Report Finished message
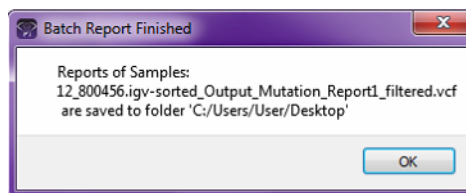
10. Click OK.

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 configuration. 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. Open the location from which you are deleting the report:

   • The Samples sub-tab on the Patient tab.

   • The Runs tab.

   • The Samples pane on the Run tab.

   • The Associated Samples pane on the Variant tab.

2. Do one of the following:

   • Select the sample or CTRL-click to select the multiple samples for which the Batch Variants report is to be generated, and then right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.

   • Select the run or CTRL-click to select the multiple runs for which the Batch Variants report is to be generated, and then right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants report.

The Batch Variants report dialog box opens. See Figure A-11 on page 329.

3. Right-click on any heading in the Input Format pane, and then choose Select Columns.

   The Select Columns dialog box opens.

Figure A-15:   Select Columns dialog box
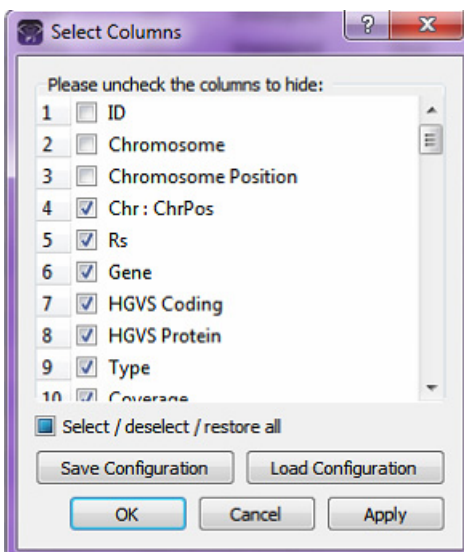
4. Click Load Configuration.

   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 A-16:   Load Configuration dialog box
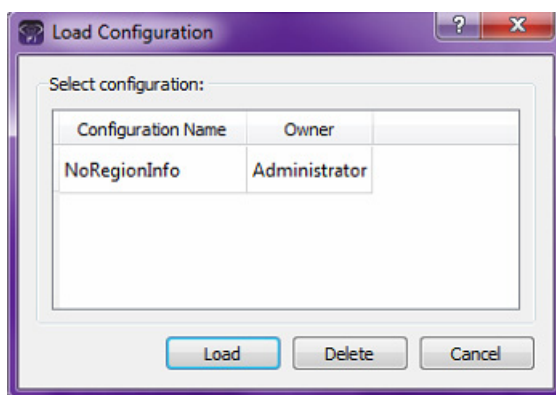
5. Select the configuration that you are deleting, and then click Delete.

   The configuration is deleted immediately. The Load Configuration dialog box remains open.

6. Click Close (x) to close the Load Configuration dialog box, and then click lose (x) to close Batch Variants Report dialog box.

Generating a Batch Coverage Report

The Batch Coverage report details the coverage information for one or more selected samples or for all the samples in a selected run or run. A separate report is saved for each sample. You can generate a Batch Coverage report from the following locations in Geneticist Assistant:

- The Samples sub-tab on the Patient tab.

- The Runs tab.

- The Samples pane on the Run tab.

- The Associated Samples pane on the Variant tab.

The report also includes coverage statistics for the panel that are calculated based on one of the following options:

- All panel samples or selected panel samples.

- Samples that have been added to the panel within the last 30 days of the current day's date, or a custom date range.

- Selected runs for the panel.

Figure A-17:   Batch Coverage Report dialog box
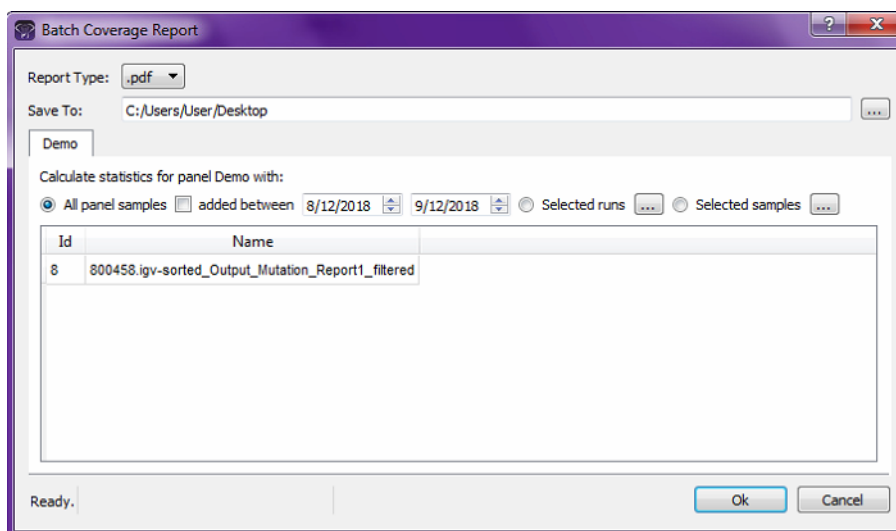
To generate a Batch Coverage report
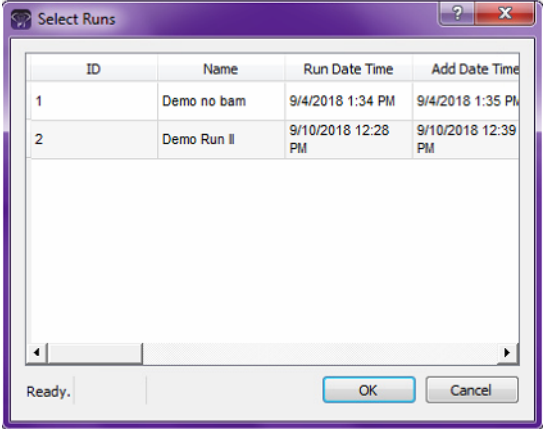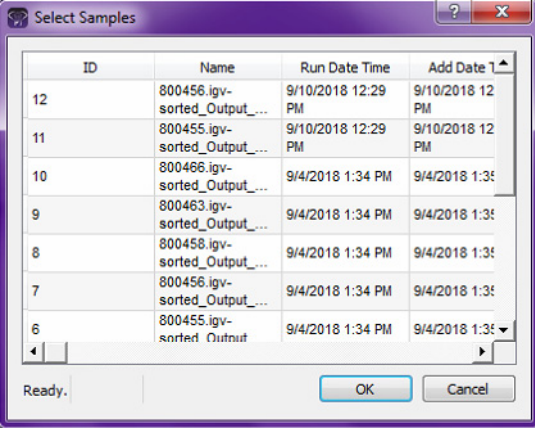
1. Open the location from which you are generating the report:

    - The Samples sub-tab on the Patient tab.

    - The Runs tab.

    - The Samples pane on the Run tab.

    - The Associated Samples pane on the Variant tab.

2. Do one of the following:

    - Select the sample or CTRL-click to select the multiple samples for which the Batch Coverage report is to be generated, and then right-click any selected sample, and on the context menu that opens, click Export > Create Batch Coverage report.

    - Select the run or CTRL-click to select the multiple runs for which the Batch Coverage report is to be generated, and then right-click any selected run, and on the context menu that opens, click Export > Create Batch Coverage report.

    The Batch Coverage report dialog box opens. See

3. Leave the report type set to the default value of .csv, or select a different report type.

4. Next to the Save To field, click the Browse button [...] to open the Save To dialog box, and then browse to and select the location in which to save the report.

5. To calculate the panel coverage statistics for the report based on all panel samples, click OK; otherwise, specify the different options (listed in the table below) by which to calculate the panel coverage statistics, and then click OK.

    After you click OK to generate the report, the Current Jobs tab opens. The tab displays the progress of calculating the coverage information for every sample that was selected for the report. A report file is saved in the specified format and the specified location.

Option	Description
All panel samples added between	To specify a custom date range for calculating the panel coverage statistics, select added between, and then change one or both dates as needed. To change the dates, you can do either of the following:
	• You can select any part of the date that you want to change, or the entire date, and then manually enter the new value. (The default date range is the last 30 days).
	• You can select any part of the date that you want to change, and then use the Up/Down arrows to change the date accordingly.

Option	Description
Selected runs	To select specific runs for calculating the panel coverage statistics, choose Selected runs, and then next to Selected runs, click the Browse button [...] to open the Select Runs dialog box. By default, all the runs for the panel are displayed in the dialog box. Select a run, or CTRL-click to select multiple runs, and then click OK. Figure A-18: *Select Runs dialog box*
Selected samples	To select only specific samples for calculating the panel coverage statistics, choose Selected samples, and then next to Selected samples, click the Browse button [...] to open the Select Samples dialog box. By default, all the samples for the panel are displayed in the dialog box. Select a sample, or CTRL-click to select multiple samples, and then click OK. Figure A-19: *Select Samples dialog box* **Note:** If the report is to be generated based on only a small number of samples, such as two to four, then, before the statistics are calculated, a Very few samples message dialog box opens, warning you of this and asking you if you want to continue. You can click Yes, or you can click Cancel to return to the Patient tab.

Creating 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 required. You can create an Action list for the following:

- Panel regions on the Coverage Regions tab.

- Variants on the Sample Variant Data sub-tab on the Sample tab.

To create an Action list

1. Do one of the following:

- On the Coverage Regions tab:

  i Select a single panel, or CTRL-click to select multiple panels.

  ii. Right-click any selected panel, and on the context menu that opens, click Add Selected Regions to Action List.

- On the Sample Variant Data sub-tab on the Sample tab:

  i Select a variant, or CTRL-click to select multiple variants.

  ii. Right-click any selected variant, and on the context menu that opens, click Add Reports > Add Selected Variants to Action List.

   The Add to Action List dialog box opens.

Figure A-20:   Add to Action List dialog box
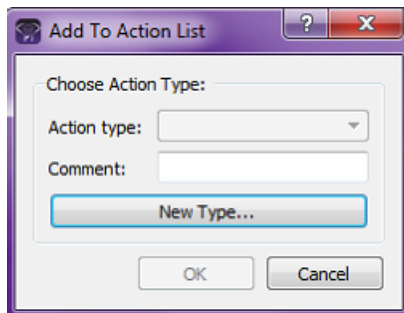
2. Do one of the following:

- If the Action Type that you are using is already available on the Action Type dropdown list, then go to Step 6.

- If this is the first time that you are creating an Action list for the pane, or if the Action Type that you want to use is not available on the Action Type dropdown list, then you must first add at least one type to the list. Go to Step 3.

3. Click New Type.

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

4. 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.

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

6. 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 you are creating an Action list for variants, then only the action types that were created specifically for variants are available.

7. 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.

8. 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 added for an Action list.

Figure A-21: Action List tab

Comparing Samples in a Run

You can compare all the samples in a run from the Runs tab, or you can compare selected samples for a run from the Run tab.

To compare samples in a run

1. Do one of the following:

   • From the Runs tab: Right-click the appropriate run on the runs tab, and on the context menu that opens, click Compare Samples.

   The Sample Comparison dialog box opens. All the samples for the selected run are displayed in the dialog box.

   • From the Run tab:

   i   Open the Run tab for the appropriate run.

   ii. CTRL-click to select the samples in the Samples pane that you are comparing,

   iii. Right-click any selected sample and on the context menu that opens, click Compare Samples.

   The Sample Comparison dialog box opens. All the selected samples for the run are displayed in the dialog box.

Figure A-22: Sample Comparison dialog box
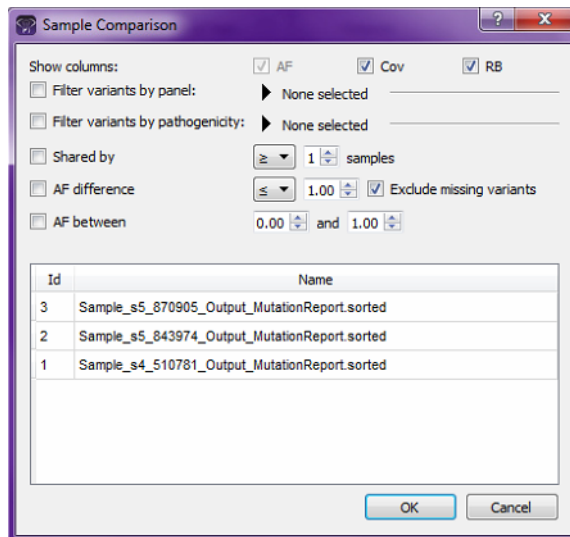
2. Set the comparison parameters.

Option	Description
Show columns	The data columns that are displayed for each sample on the Sample Comparison tab. Allele Frequency (AF) is always displayed and you cannot change this option. By default, Coverage (Cov) is selected.
Filter variants by panel	Limits the variants that are displayed don the Sample Comparison tab to only those variants that are contained within the selected panel.
Filter variants by pathogenicity	Limits the variants that are displayed on the Sample Comparison tab to only those variants with the selected pathogenicity.
Shared by	Limits the variants are displayed on the Sample Comparison tabl to only those variants that meet the required number of samples as specified by the operator that is selected on the dropdown list (>, <, or =) and the number of samples. If a variant is found a number of samples that does meet this criteria, then the variant is not displayed in the results.
AF difference	Limits the variants are displayed on the Sample Comparison tabl to only those variants that meet the required number of samples as specified by the operator that is selected on the dropdown list (>, <, or =) and the AF difference value. If a variant is found a number of samples that does meet this criteria, then the variant is not displayed in the results.
• Exclude missing variants	Selected by default. Regardless of the allele frequency difference, always display variants that are *not* found in one or more samples.
AF between	Limits the variants that are displayed on the Sample Comparison tab to only those variants with an allele frequency (AF) that falls within the indicated range in at least one of the compared samples.

3. Click OK.

The Sample Comparison dialog box closes. The Sample Comparison results are displayed on the Sample Comparison tab. The far right side of the tab shows the sample-specific information for each variant, with each sample identified as 1, 2, and so on. If you hover your cursor over the column heading, then the full sample name is displayed. The left side of the tab shows the variant information.

Figure A-23:   Sample Comparison tab

Adding a Variant Interpretation

A variant interpretation is a comment amount or a description of a variant. You can add interpretations to the variants in a patient sample from the Variants sub-tab on the Patient tab or from the Sample Variant Data sub-tab on the Sample tab.

To add a variant interpretation

1. Open the Variant sub-tab on the Patient tab or the Sample Variant Data sub-tab on the Sample tab.

2. Select a single variant, or CTRL-click to select multiple variants and then right-click to open the sub-tab context menu.

3. On the context menu, select Edit Variant and then if:

   - A single variant is selected, select Add Single Variant Interpretation to open the Add Single Variant Interpretation dialog box. See Figure A-24 below.

   - Two or more variants are selected, select Add Compound Variant Interpretation to open the Add Compound Variant Interpretation dialog box. All the selected variants are displayed in the Included Variants (left) pane when the dialog box opens. See Figure A-25 on page 343.

Figure A-24: Add Single Variant Interpretation dialog box
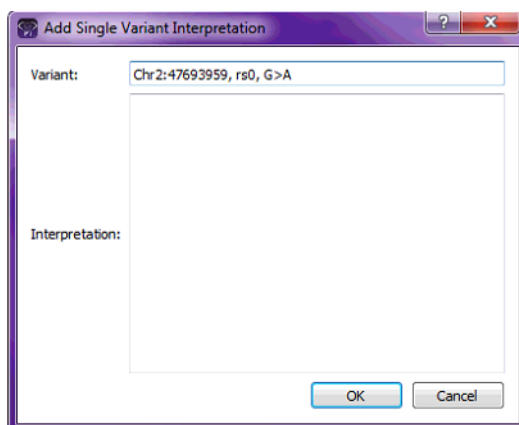
Figure A-25:   Add Compound Variant Interpretation dialog box
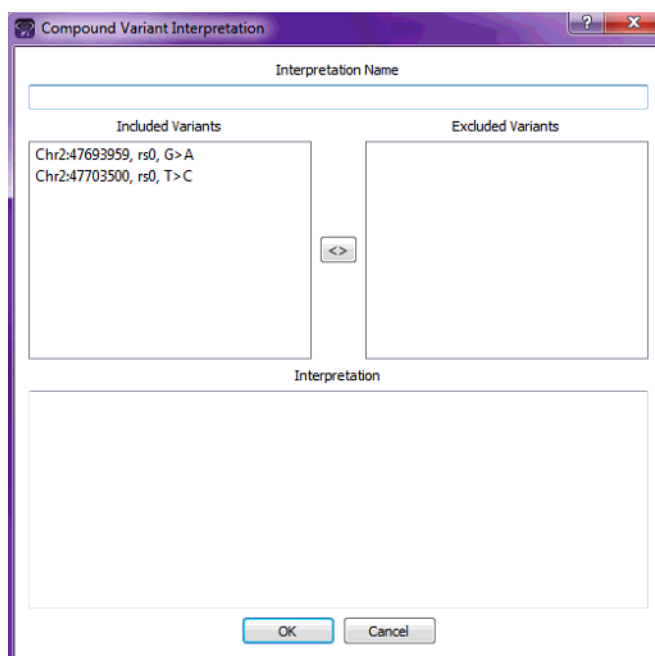
4. Do one of the following:

    •   For a single variant, in the blank Interpretation pane, enter the appropriate comment or description, and then click OK.

        The Add Single Variant Interpretation dialog box closes. The interpretation is displayed in the Interpretation column on the Variants sub-tab for the Patient tab.

    •   For two or more variants, in the Interpretation Name field:

        i   Enter an identifying name for the interpretation.

        ii. Leave all the variants as-is in the Included Variants pane, or optionally, select one or more included variants (CTRL-click to select multiple variants), and then click the Move arrow to move these selected variants to the Excluded Variants (right) pane.

        iii. Optionally, in the Interpretation field, enter a description of the interpretation.

        iv. Click OK.

        The Add Compound Variant Interpretation dialog box closes. If you open the Sample tab from the Patient tab, then you can see the interpretation displayed in the Compound Variant Interpretations pane on the Sample Data sub-tab. You can double-click an interpretation that is displayed in the Compound Variant Interpretations pane to open a Compound Variant Interpretation sub-tab that displays the variants that are included for the interpretation. See "Compound Variant Interpretation Sub-Tab" on page 240.

Appendix A
Common Procedures

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 Regions tab or for variants on the 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 tab and the Coverage Regions sub-tab on the Panel Statistics 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 Geneticist Assistant. 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

Chemistry

Refers to the chemistry that was used to prepare the samples for sequencing. Values can be specified through the Add/Edit Object function in Geneticist Assistant.

CIVic Data pane

Found on the Variants tab. Displays information for the variant from the CIViC (Clinical Interpretations of Variants in Cancer) database.. See https://civicdb.org/home.

Client Reference Directory

Contains the information for the reference that was used for aligning data and/or detecting variants. If the Geneticist Assistant client and server reside on the same computer, then you can set the Client Reference directory and the Server Reference directory to the same directory.

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/.

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 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 user-specified data.

Custom report template

Specifies not only the data that a Sample PDF report is to contain, but also the layout of the report. You build a custom report template in the Report Designer.

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

Ethnicity

The ethnicity (for example, Caucasian/Non-Hispanic) for a patient in your Geneticist Assistant database. Values can be specified through the Add/Edit Object function in Geneticist Assistant.

Exome Aggregation Consortium pane

Found on the Variant tab. Displays the information for the variant from the Exome Aggregation Consortium database for selected global sequencing projects. See http://exac.broadinstitute.org/.

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.

gnomAD Data pane

Found on the Variant tab. Displays information for the variant from the gnomAD (Genome Aggregation Database) database. See http://reusabledata.org/gnomad.html.

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%.

I

Instrument

Refers to the instrument that was used to sequence the samples. Values can be specified through the Add/Edit Object function in Geneticist Assistant.

Interpretation

A description or explaining comment for a variant. A variant can have one or more single interpretations associated with it, or it can be part of a compound interpretation, which means that the interpretation describes or explains multiple variants.

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 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.

O

Object

Holds a value or variable that can be applied for carrying out runs in Geneticist Assistant.

OMIM Data pane

Found on the Variants tab. Displays the information for the variant from the OMIM (Online Mendelian Inheritance in Man) database. See http://www.omim.org/.

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 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.

Phenotype

The phenotype for a patient. Values can be specified through the Add/Edit Object function in Geneticist Assistant.

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.

Run

Refers to loading a BAM file and a sorted VCF file or just a sorted 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.

Run Status

The status for a run. Default values are New, QC Passed, Reviewed, and Complete. Additional values can be specified through the Add/Edit Object function in Geneticist Assistant.

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.

Server Reference Directory

The directory on the Geneticist Assistant database server that hosts the reference files. If the Geneticist Assistant client and server reside on the same computer, then you can set the Client Reference directory and the Server Reference directory to the same directory.

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 Regions, Panel Statistics, Sample, Variant, and Action List.

Standard user

A Geneticist Assistant user who has neither Administrator privileges nor Staff status.

Subpanel

Contains a fixed number of regions that are contained within a selected panel. To verify coverage for a smaller number of regions that are within the panel that has been selected for a run, you can use a subpanel.

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 Sample Variant Data sub-tab on the Sample 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 Geneticist Assistant 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.

W

Web user

A person who can log in and submit information to the Geneticist Assistant customer portal.

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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