

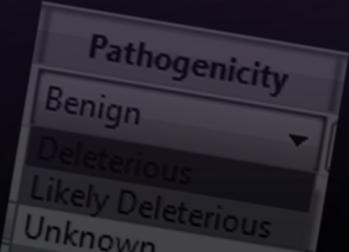
GeneticistAssistant™

NGS Interpretative Workbench

User Manual



SOFTGENETICS®



SOFTGENETICS®

Release Information

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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 the Geneticist Assistant application efficiently and effectively.

Using the manual

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

Special information about the manual

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



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

Conventions used in the manual

The *Geneticist Assistant User's Manual* uses the following conventions:

- Information that can vary in a command—variable information—is indicated by alphanumeric characters enclosed in angle brackets; for example, <Gene ID>. Do not type the angle brackets when you specify the variable information.
- A new term, or term that must be emphasized for clarity of procedures, is *italicized*.
- Page numbering is “online friendly.” Pages are numbered from 1 to x, *starting with the cover* and ending on the last page of the index.



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

- This manual is intended for both print and online viewing.
 - If information appears in [blue](#), it is a hyperlink. Table of Contents and Index entries are also hyperlinks. Click the hyperlink to advance to the referenced information.

Assumptions for the manual

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

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

Organization of the manual

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

- [Chapter 1, “Getting Started with Geneticist Assistant,” on page 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 the application, the main menu, the toolbar, the tabs display area, and the status bar.
- [Chapter 2, “Geneticist Assistant Setup and Maintenance,” on page 47](#) 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. Finally, it explains how to back up the Geneticist Assistant database.

- [Chapter 3, “Geneticist Assistant User Management,” on page 69](#) 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 83](#) details the management of your Geneticist Assistant panels, which includes, adding, modifying and deleting panels, and reviewing panel statistics, and adding, modifying, and deleting subpanels.
- [Chapter 5, “Managing Patients and Runs,” on page 105](#) 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 155](#) 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 179](#) 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 215](#) 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 245](#) 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 267](#) details the generation of the six standard reports that are available in Geneticist Assistant. It also details how to generate a custom report, which includes only the data that you select.
- [Chapter 11, “Geneticist Assistant Tools,” on page 277](#) details the various tools that are available in Geneticist Assistant to assist you with setting up, running, and analyzing sample data.
- The [Glossary on page 291](#) provides a detailed definition of specialized terms for Geneticist Assistant.

Preface

Chapter 1

Getting Started with Geneticist Assistant

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

This chapter covers the following topics:

- [“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.](#)
- [“Installing the Geneticist Assistant Database Server on a Linux OS” on page 19.](#)
- [“Starting and Logging in to Geneticist Assistant” on page 21.](#)
- [“Upgrading Geneticist Assistant” on page 27.](#)
- [“The Geneticist Assistant Main Window” on page 28.](#)
- [“Setting Display Preferences in the Geneticist Assistant Main Window” on page 31.](#)
- [“Searching the Geneticist Assistant Database” on page 44.](#)

Chapter 1
Getting Started with Geneticist Assistant

Geneticist Assistant System Recommendations

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

Table 1-1: Geneticist Assistant Recommended Hardware Specifications

Client and/or Server	
Operating System	<ul style="list-style-type: none"> 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	<ul style="list-style-type: none"> 8 GB (Client) 4 GB (Server) Minimum: 2 GB for client and server
Hard Drive	<ul style="list-style-type: none"> Solid State Drive 100 GB (Minimum)
Note: Virtual Machines are supported.	

Table 1-2: Geneticist Assistant Data Specifications

Database	Client
<ul style="list-style-type: none"> 4 GB disk space (Minimum) 	<ul style="list-style-type: none"> 50 GB space for reference and annotation data
<ul style="list-style-type: none"> Starts at 15 MB and grows slowly 	<ul style="list-style-type: none"> BAM files - Ca. the same size as raw FASTQ data
<ul style="list-style-type: none"> Backup with mysqldump 	
<ul style="list-style-type: none"> Low IOPS 	

Table 1-3: Geneticist Assistant Software Details

Server	Client
<ul style="list-style-type: none"> Apache 2.2 (Latest) 	<ul style="list-style-type: none"> Qt 4.84
<ul style="list-style-type: none"> mod_wsgi (Latest) 	<ul style="list-style-type: none"> Bamtools 2.1.1
<ul style="list-style-type: none"> OpenSSL (Latest) 	<ul style="list-style-type: none"> Zlib 1.2.8
<ul style="list-style-type: none"> MySQL 5.6 (Latest) 	<ul style="list-style-type: none"> Qjson 2.1
<ul style="list-style-type: none"> Python 2.7 (Latest) <ul style="list-style-type: none"> Django 1.6 (Latest) South (Latest) Setuptools (Latest) MySQLdb (Latest) 	<ul style="list-style-type: none"> OpenSSL 0.9.7

Installing the SoftGenetics License Server

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



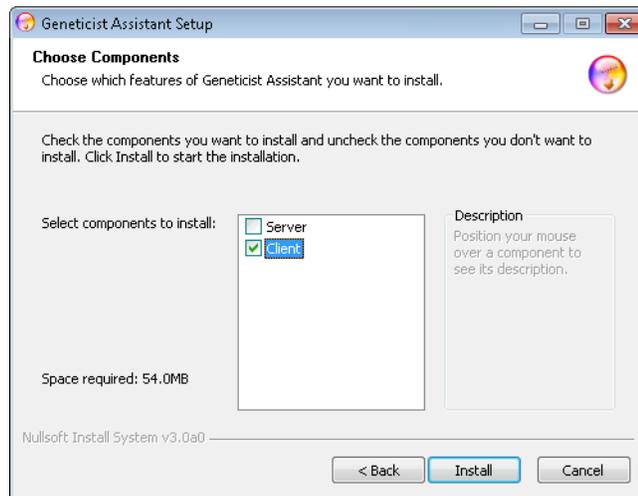
If you are running a trial version of Geneticist Assistant, then you do not need to set up the SoftGenetics license server. You can simply indicate that you are running a trial version after you log in to Geneticist Assistant.

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. When the installer first opens, Server is *not* selected and Client is selected.

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

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



When you install Geneticist Assistant, you are prompted to create the Administrator role for the installation. By default, the Username is set to Administrator, but you can always enter a different value. The password cannot contain any spaces. If you intend to implement additional password requirements, then the password must also adhere to these planned requirements. [Figure 1-2 on page 18](#) shows the requirements that you can specify for user passwords after Geneticist Assistant is installed. (See [“To specify the Geneticist Assistant password requirements” on page 63.](#))

Chapter 1
Getting Started with Geneticist Assistant

Figure 1-2: User password requirement options

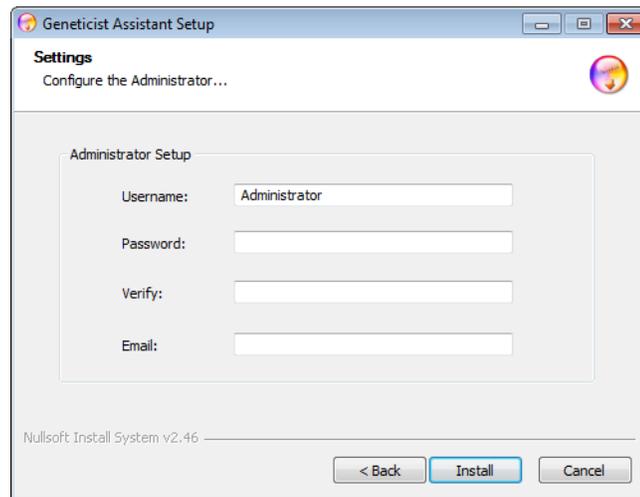


A dialog box titled "Password Requirements" with a "Reset Defaults" button at the bottom. It contains five rows of settings, each with a label on the left and a numeric input field on the right:

Requirement	Value
Length:	8
Upper Case:	1
Lower Case:	1
Digits:	1
Special Characters:	0

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

Figure 1-3: Administrator Setup dialog box



A window titled "Geneticist Assistant Setup" with a "Settings" sub-header and the text "Configure the Administrator...". It contains an "Administrator Setup" section with four input fields:

- Username: Administrator
- Password: (empty)
- Verify: (empty)
- Email: (empty)

At the bottom, there are three buttons: "< Back", "Install", and "Cancel". The footer text reads "Nullsoft Install System v2.46".

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



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

Installing the Geneticist Assistant Database Server on a Linux OS

To install Geneticist Assistant database server on a Linux OS, you must do the following:

- Create the Geneticist Assistant database.
- Set up the Geneticist Assistant database server.
- Create and migrate the initial database contents.
- Configure Apache2.
- Test the installation.



You must still install the Geneticist Assistant client on a Windows OS. See “Installing Geneticist Assistant on a Windows OS” on page 17.

To create the Geneticist Assistant database

1. If it is not already installed, install OpenSSH.
2. Verify that python2.7 is installed.
3. Install the following components in the order indicated:

Component	Installation
apache2	sudo apt-get install apache2
libapache2-mod-wsgi	sudo apt-get install libapache2-mod-wsgi
mysql	sudo apt-get install mysql-server mysql-client
pip	sudo apt-get install python-pip
Django 1.6.11	sudo pip install Django==1.6.11
South	sudo pip install south
python-mysqldb	sudo apt-get install python-mysqldb

4. Create the database, and then replace the username and password with the password that you created during the mysql-server setup.



*There is no space after -u and -p:
mysql -u<username> -p<password> -e "create database softgenetics"*

To set up the Geneticist Assistant database server

1. Download the Geneticist Assistant database server.



Contact SoftGenetics for the most recent version.

2. Extract the Geneticist Assistant server.
3. Move the Geneticist Assistant server to the installation location:
`sudo mv ga_server_<version number> /opt/`
4. Change the permissions:
`sudo chown -R www-data:www-data /opt/ga_server_<version number>/`
5. Change the directory: `cd /opt/ga_server_<version number>`
6. Edit the settings: `sudo vi ga/settings.py`, and then in the DATABASES section, configure the mysql user and password that you created during the mysql-server setup.

To create and migrate the initial database contents

1. Create the initial database contents: `python manage.pyc syncdb`, and then configure the administrator username, password, and email address for Geneticist Assistant.
2. Migrate the database: `python manage.pyc migrate`

To configure Apache2

1. To set up the Install/Create keys:
`sudo openssl req -x509 -nodes -days 3650 -newkey rsa:2048 -keyout /etc/apache2/server.key -out /etc/apache2/server.crt`
2. Configure Apache2:
 - `sudo cp softgenetics.conf /etc/apache2/site-available/`
 - `sudo a2ensite softgenetics.conf`
 - `sudo a2enmod ssl`
 - `sudo service apache2 restart`

To test the installation

Test the connection to the database server from a client with the following credentials:

- Server: `https://<hostname>`
- Username: `<created during install>`
- Password: `<created during install>`

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. After Geneticist Assistant is installed and you open Geneticist Assistant for the first time, you must specify the URL for the database server.

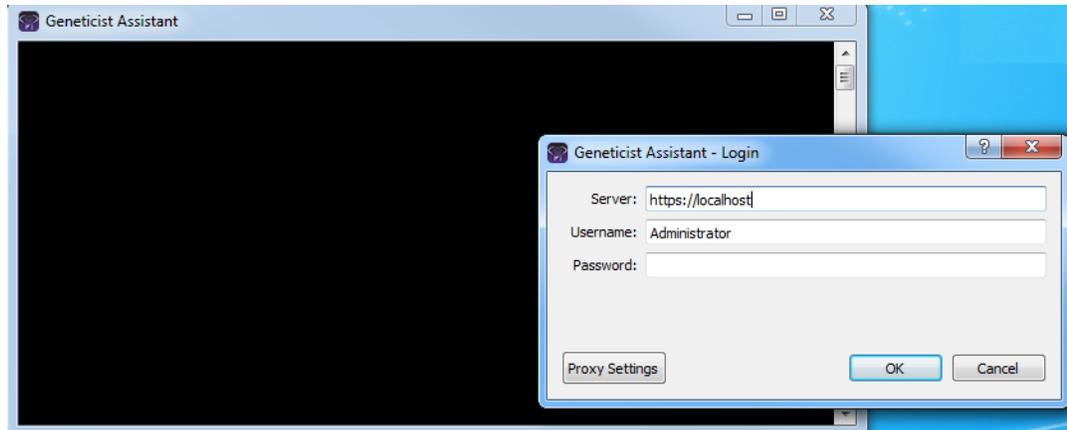
Figure 1-4: Geneticist Assistant desktop icon



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

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

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



Before you log in to Geneticist Assistant, you can click Proxy Settings to open the Proxy Settings tab on the Settings dialog box. If your organization uses a proxy server for external connections, then to view data from the external databases OMIM or LOVD in Geneticist Assistant, you must configure the settings for the proxy server on this tab. See [“To configure proxy server settings”](#) on page 66.

Chapter 1
Getting Started with Geneticist Assistant

2. If this is the *first* time that you are logging in to Geneticist Assistant, then do the following; otherwise, go directly to [Step 4](#).
 - In the Server field, enter the URL for the Geneticist Assistant database server:

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

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



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

3. Continue to one of the following:
 - If you are running a trial version of Geneticist Assistant, continue to [“To run a trial version of Geneticist Assistant”](#) on page 23.
 - If you are running a purchased version of Geneticist Assistant, continue to [“To run a purchased version of Geneticist Assistant”](#) on page 24.
4. If needed, in the Username field, enter your username, and then in the Password field, enter your password, and then click OK. One of the following happens:
 - If your login is successful, then the Geneticist Assistant Login dialog box closes, “Success” is displayed in the Debug window, and the Geneticist Assistant main window opens. The Debug window remains open.
 - If your login is not successful, then the reason for the failure is displayed in the Debug window. If you are unable to resolve the login issues based on the messages in the Debug window, contact SoftGenetics for assistance.



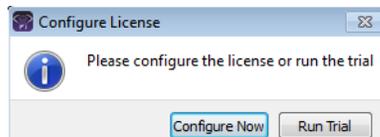
The Debug window displays the results after each step that you carry out in Geneticist Assistant. You must leave this window open for the duration of your session. If you close it, then you also close Geneticist Assistant.

5. 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 *always* opens.

Figure 1-6: *Configure License message*

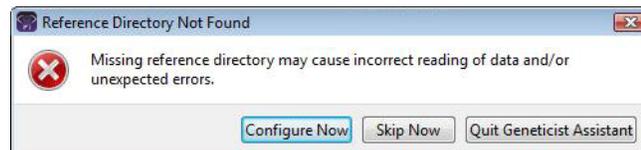


1. Click Run Trial.

The Configure License message closes. Two results are possible.

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

Figure 1-7: *Reference Directory Not Found message*



- If this is *not* the first time that you are running the trial version, and the reference directory has already been configured, then the Geneticist Assistant main window opens. Continue to [“The Geneticist Assistant Main Window”](#) on page 28.
2. Do one of the following:
 - To configure the reference directory now, click Configure Now to open the Settings dialog box, and then continue to [“To configure the reference directory”](#) on page 26.
 - To configure the reference directory at a later date, click Skip Now.

The Reference Directory Not Found message closes and the Geneticist Assistant main window opens. Continue to [“The Geneticist Assistant Main Window”](#) on page 28.

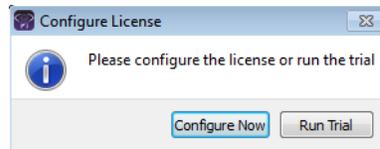


For detailed information about configuring the required reference directory at a later date, see [“Specifying your Geneticist Assistant Settings”](#) on page 53.

To run a purchased version of Geneticist Assistant

If you are running a purchased version of Geneticist Assistant, then the first time that you run the application, a 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.

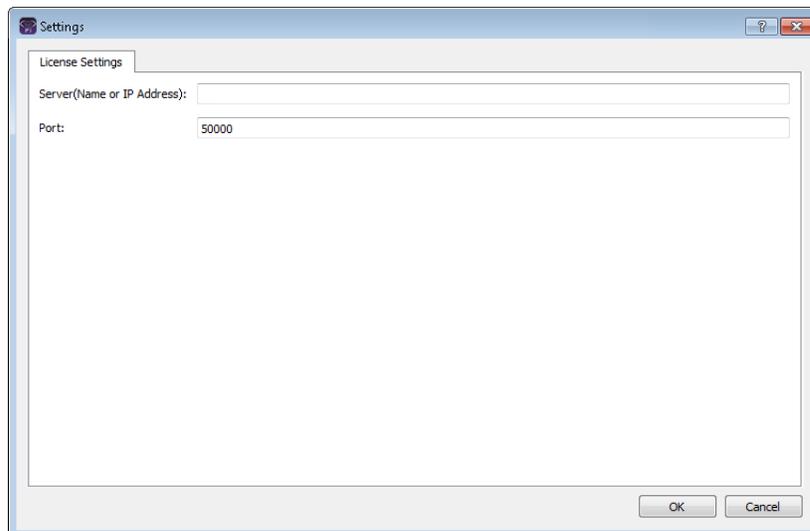
Figure 1-8: Configure License message



1. Click Configure Now.

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

Figure 1-9: Settings dialog box



2. In the server name, enter the fully qualified domain name (FQDN) for the server (where the license server manager is installed), or its IP address.



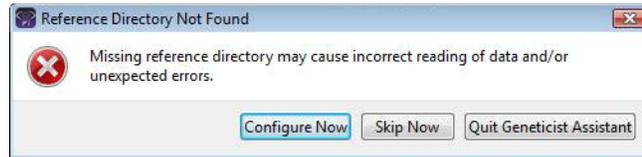
If the SoftGenetics license server is running on the same computer on which Geneticist Assistant is running, then you must enter 127.0.0.1.

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

4. Click OK.

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

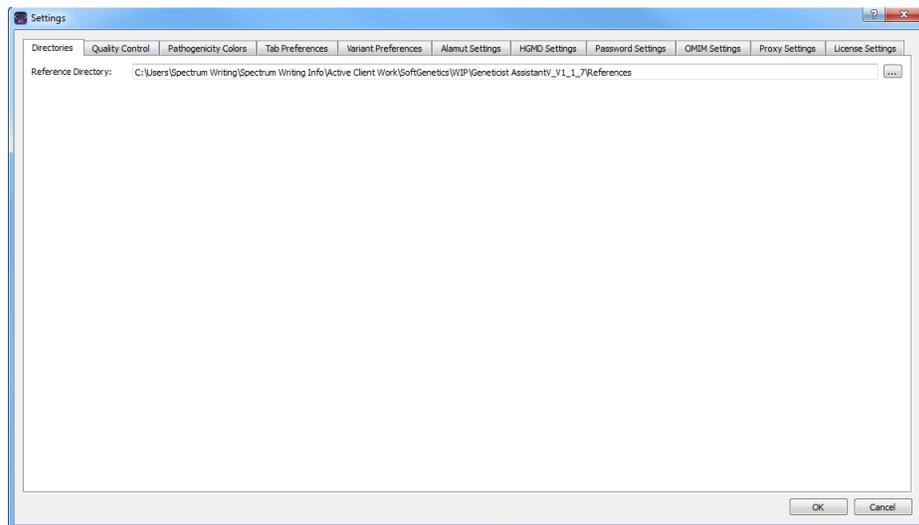
Figure 1-10: Reference Directory Not Found message



5. Do one of the following:

- To configure the reference directory now, click Configure Now to open the Settings dialog box, and then continue to [“To configure the reference directory” on page 26.](#)

Figure 1-11: Settings dialog box, Directories tab



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

The Reference Directory Not Found message closes and the Geneticist Assistant main window opens. Continue to [“The Geneticist Assistant Main Window” on page 28.](#)



For detailed information about configuring the required reference directory at a later date, see [“Specifying your Geneticist Assistant Settings” on page 53.](#)

To configure the reference directory

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



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

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



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

2. Click the Browse button  next to the Reference Directory field, and then browse to and select the Reference directory.
3. Click OK to close the Settings dialog box and return to the Geneticist Assistant main window.
4. Continue to [“The Geneticist Assistant Main Window” on page 28.](#)

To log out of Geneticist Assistant

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

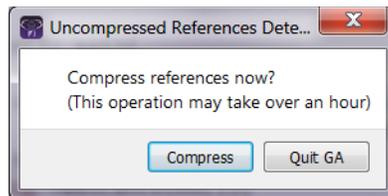


The Debug window displays the results after each step that you carry out in Geneticist Assistant. You must leave this window open for the duration of your session. If you close it, then you also close Geneticist Assistant.

Upgrading Geneticist Assistant

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

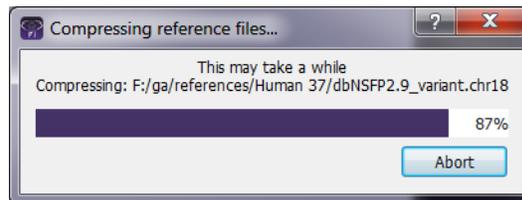
Figure 1-12: Uncompressed References Detected message dialog box



1. Click Compress.

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

Figure 1-13: 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.

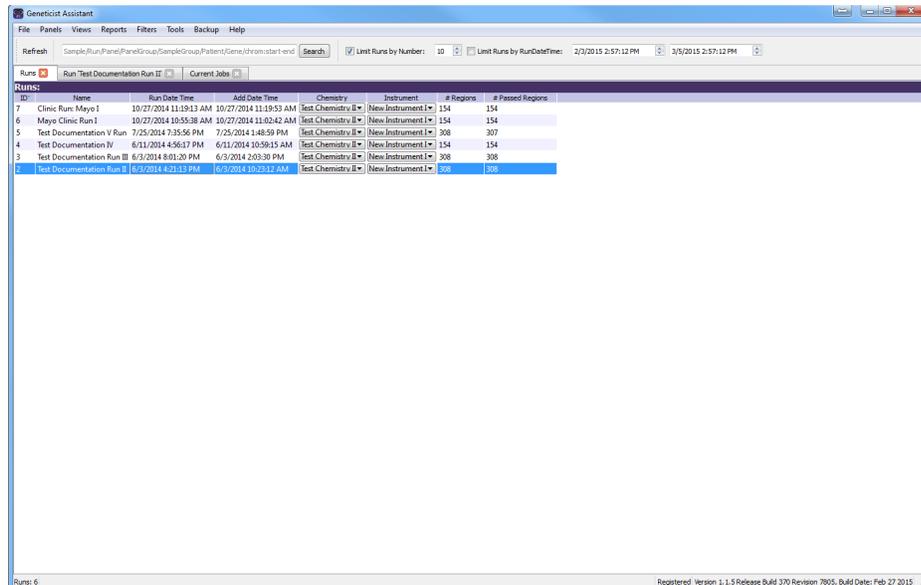
2. After the Compression Progress dialog box indicates that the compression is 100% complete, click OK.

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 the Geneticist Assistant application.

Figure 1-14: Geneticist Assistant main window



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

Title bar

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

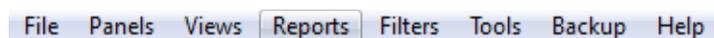
Figure 1-15: Geneticist Assistant title bar



Main menu

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

Figure 1-16: Geneticist Assistant main menu



Toolbar

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

- You use the Refresh option to manually refresh a tab display after the tab opens in the main window and data is loaded, or after adding, modifying, and/or deleting data on a tab. See “[Main window tabs](#)” below.
- You use the Search feature to search for samples, runs, panels, panel groups, sample groups, genes, or variants in your Geneticist Assistant database. See “[Searching the Geneticist Assistant Database](#)” on page 44.

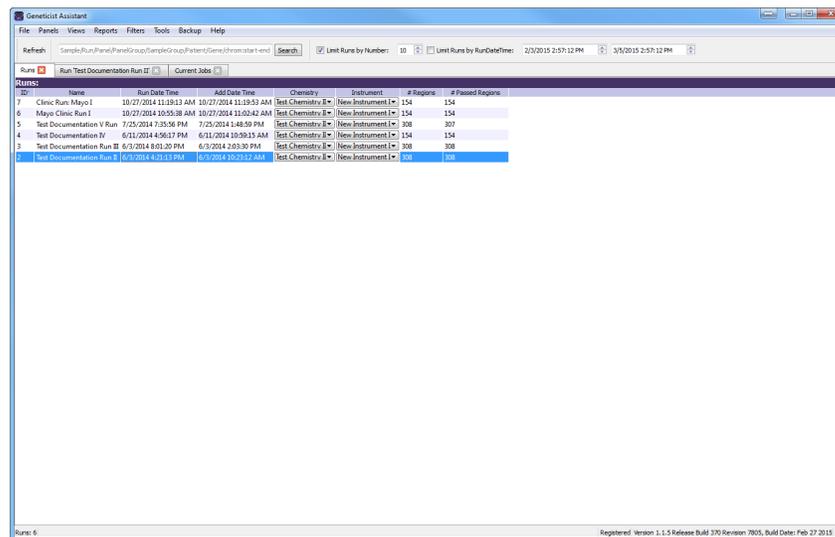
Figure 1-17: Geneticist Assistant main window toolbar



Main window tabs

When the Geneticist Assistant main window first opens, two tabs—Runs and Current Jobs—are displayed in the window. The Runs tab is the open tab. A *run* refers to loading a BAM 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 1-18: 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.

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- 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 1-19: Geneticist Assistant main window, Status bar



Setting Display Preferences in the Geneticist Assistant Main Window

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

- Navigate multiple open tabs/minimized window. See [“To navigate multiple open tabs/ Navigate tabs with a minimized main window”](#) on page 32.
- Close multiple tabs in a single step. See [“To close multiple tabs in a single step”](#) on page 32.
- Show/hide the toolbar for a tab. See [“To show/hide the toolbar for a tab”](#) on page 32.
- Show/hide a tab pane. See [“To show/hide a tab pane”](#) on page 32.
- Filter the data that is displayed in a 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 37.
- Manage tab pane configurations. See [“To manage tab pane configurations”](#) on page 39.
- Change the widths of a data columns. See [“To change the widths of data columns”](#) on page 42.
- Rearrange the order of data columns. See [“To rearrange the order of data columns”](#) on page 42.
- Change the sort order of data. See [“To change the sort order of data”](#) on page 42.
- Rename column headers. See [“To rename column headers”](#) on page 43.
- 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 43.

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 the application. 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 58.

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 show/hide the toolbar for a tab

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

To show/hide a tab pane

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

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 Filters table to display all the data (fields) for a selected tab on a pane. You specify filters in a Filters table to limit the display in a tab pane to only the data (fields) that meet specific criteria. You can specify the filters for the data that is displayed in a tab pane on a one time only basis, or you can save the filters to a template, and then load the template to filter the data that is displayed in the tab pane anytime as needed.

1. Open the appropriate tab.

- 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.

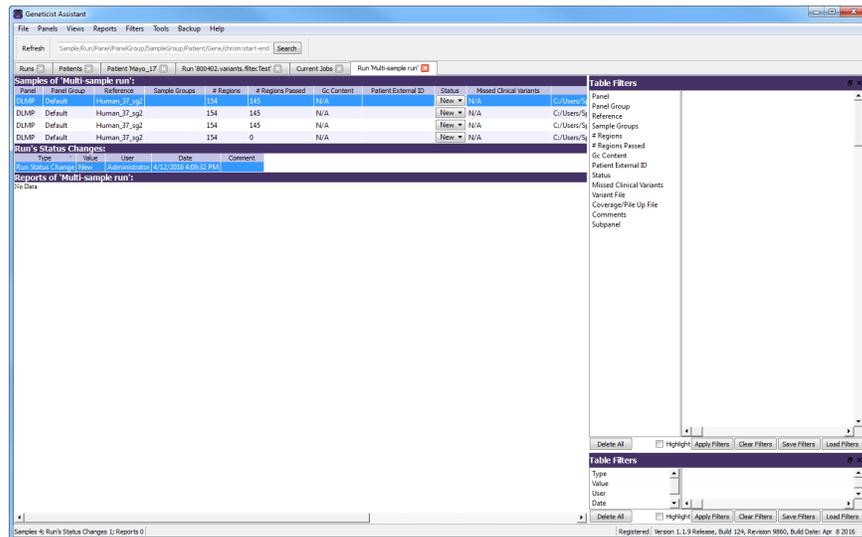
The Filters table opens to the right of the tab. The table lists all the fields for the selected tab pane.

- Repeat [Step 2](#) as many times as needed to open multiple Filters tables for a tab pane.



The remainder of this procedure describes how to set up new filters for filtering data. Optionally, you can click Load Filters to open the Load Filter Template dialog box and select from a list of saved filter templates. You can also delete saved filter templates. See “To delete a saved filter template” on page 37.

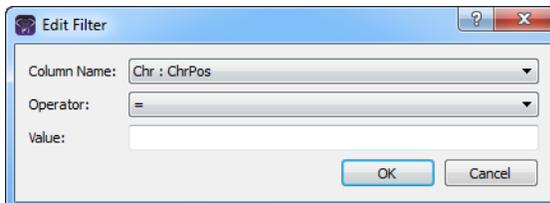
Figure 1-20: Filters tables opened for a tab



- 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.

Figure 1-21: Edit Filter dialog box





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



If you are filtering data based on a date/time field, then for additional information, see [“To filter data based on a date/time field”](#) on page 36.

- 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.
>=	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.

- Enter the filter value.

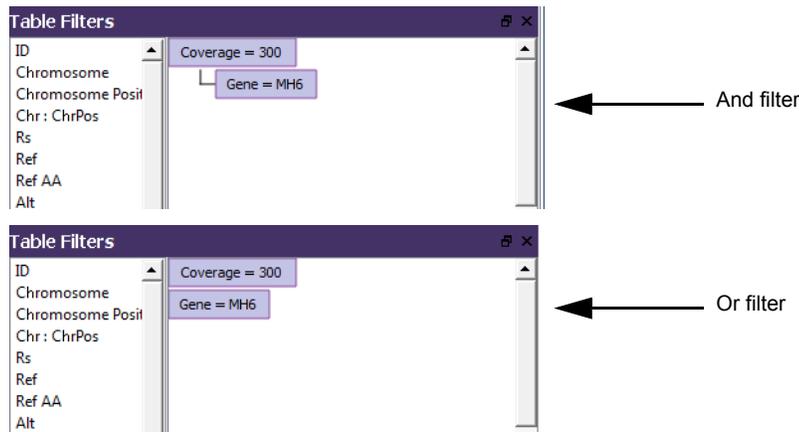
7. Click OK.

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

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

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

Figure 1-22: And and Or filters



9. Do any of the following as needed:

- To highlight the results that pass the filtering criteria instead of removing data that fails the filtering criteria, select Highlight.
- Click Apply Filters to immediately apply the filters and view the results onscreen. (“*Filters Applied” is displayed in the title of the affected tab panes.)



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

- Click Clear Filters to remove the filters that have been applied for the display for all panes on a tab in a single step. The filters remain displayed in the Filter table.

- Right-click any filter in the filter pane, 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 save the edited filters. You can then click Apply Filters to immediately apply the edited filters and view the updated results onscreen.
- Right-click the filter in the filter pane, and on the context menu that opens, click Remove to delete the filter. You can then click Apply Filters to immediately remove the filters from the display and view the updated results onscreen.
- Click Save Filters to open the Save Filter Template dialog box and save your filter settings. You can use this saved Filter Settings template to filter the data on this specific tab pane for another project based on the settings in the template.



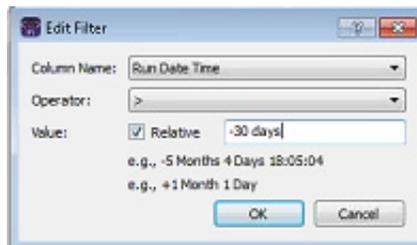
You can delete saved filter templates. See [“To delete a saved filter template”](#) below.

- Click Delete Filters to remove all filters that have been added for each tab in a single step and delete all the filters from the Filter table.
10. After you are done filtering the data, you can click Close (x) in the upper right corner of a Filter table to close it.

To filter data based on a date/time field

If you are filtering data based on a date/time field, then an additional option, Relative, 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 \leq operator, select Relative and then in the Relative field, enter -30 days. (You must enter the minus sign.)

Figure 1-23: Edit Filter dialog box for a date/time field



To delete a saved filter template

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

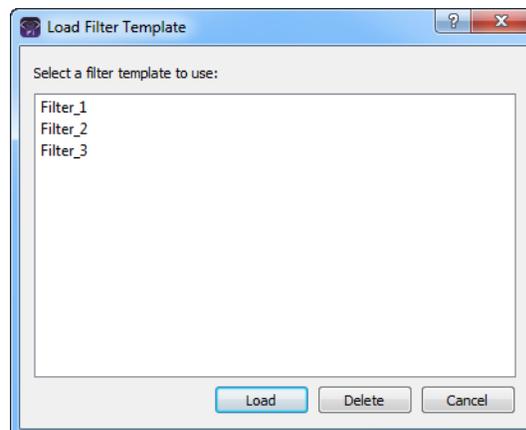
1. Open the appropriate tab.
2. On the Geneticist Assistant main menu, click Filters > <Tab Pane>. For example, if you clicked Filters pane with the Sample tab open, then the menu options would be Filters > Sample's Variants, Filters > Sample's Status Changes, and so on.

The Filters table opens to the right of the tab. The table lists all the fields for the selected tab pane. See [Figure 1-20 on page 33](#).

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 1-24: 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 can specify which columns to show in a tab pane and which data columns to hide 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 have made 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 added for the view 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 data columns are hidden from view in the 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 (that is, which columns were shown and which columns were hidden) are restored, and a completely filled checkbox is displayed for the Select/deselect/restore option.

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 each configuration displayed next to the configuration.

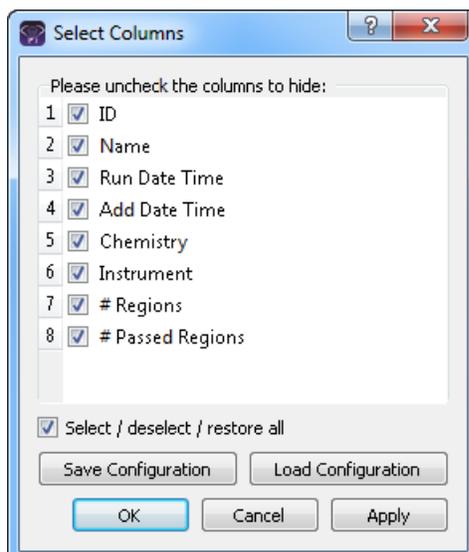


Compact displays the data columns in the pane in a more condensed view. To toggle the display, click Compact as needed.

2. Click Manually.

The Select Columns dialog box opens.

Figure 1-25: Select Columns dialog box



3. Clear the options for the columns that are to be hidden from view in the pane, and leave the options for the columns that are to be shown selected.
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 42.

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/modify a pane configuration”](#) below.
- [“To delete a pane configuration”](#) on page 41.

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/modify a pane configuration

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

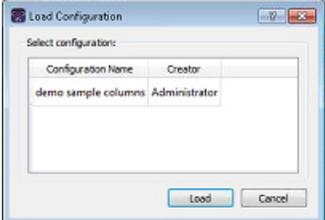
1. Right-click the tab pane anywhere, including the column headers, and on the context menu that opens, click Load Columns.

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:

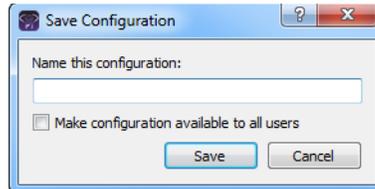
Option	Description
To create an entirely new pane configuration	<ol style="list-style-type: none"> 1. Click Manually. The Select Columns dialog box opens. By default, when the dialog box first opens for a tab pane, the columns that are currently displayed are selected, and Select/deselect/restore is either selected or completely filled. See Figure 1-25 on page 38. 2. Continue to Step 3.
To create a new pane configuration by modifying an existing configuration	<ol style="list-style-type: none"> 1. Click Manually. The Select Columns dialog box opens. See Figure 1-25 on page 38. 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. <i>Figure 1-26: Load Configuration dialog box</i> 

3. Clear the options for the columns that are to be hidden from view in the pane, and select the options for the columns that are to be displayed.
4. Click Apply to update the column display.

5. Click Save Configuration.

The Save Configuration dialog box opens.

Figure 1-27: Save Configuration dialog box



6. Enter a name for the configuration.



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

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.

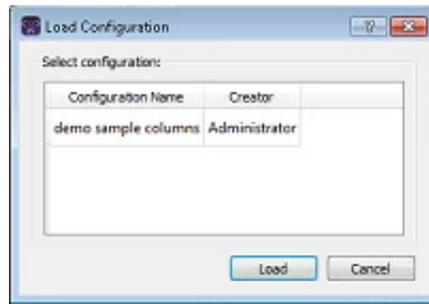
1. Right-click the tab pane anywhere, including the column headers, and on the context menu that opens, click Load Columns.

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 [Figure 1-25 on page 38](#).
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 1-28 on page 42](#).

Figure 1-28: 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 37.

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 modify 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 273.

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 1-29: Geneticist Assistant main window toolbar

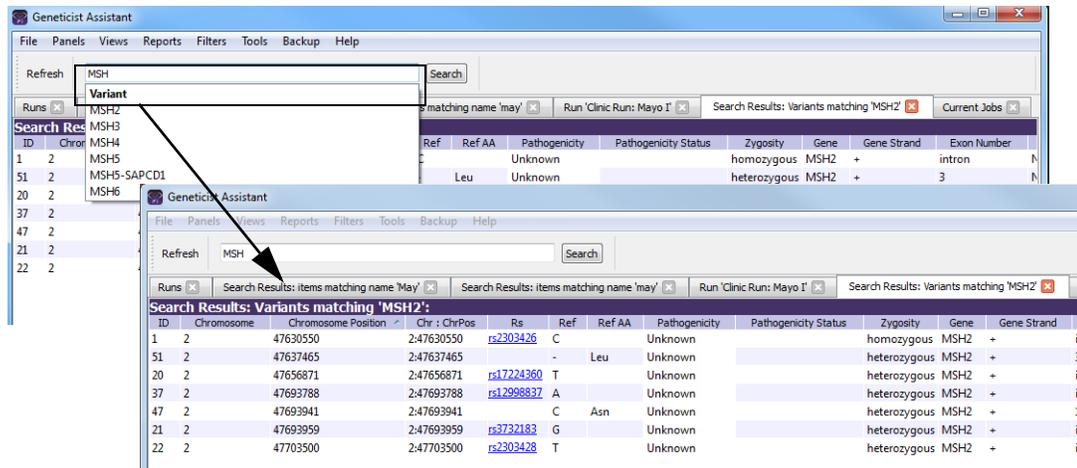


Important points to note about a search are the following:

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

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

Figure 1-30: Search feature and results example



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

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

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

Chapter 1
Getting Started with Geneticist Assistant

Chapter 2

Geneticist Assistant Setup and Maintenance

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

This chapter covers the following topics:

- [“Managing Objects” on page 49.](#)
- [“Specifying your Geneticist Assistant Settings” on page 53.](#)
- [“Backing up the Geneticist Assistant Database” on page 68.](#)

Chapter 2
Geneticist Assistant Setup and Maintenance

Managing Objects

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

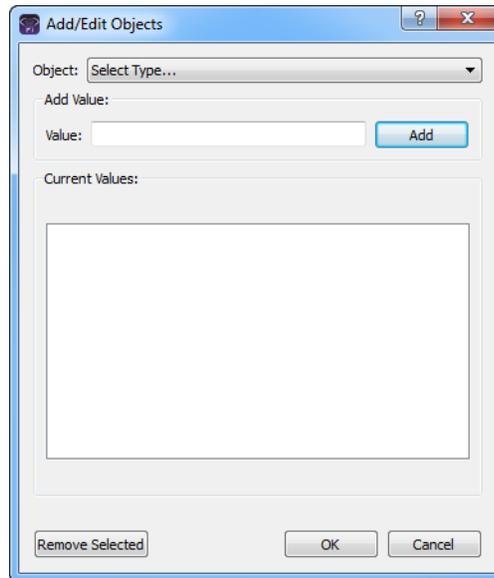
Object	Description
Chemistry	Refers to the chemistry that was used to prepare the samples for sequencing.
Instrument	Refers to the instrument that was used to sequence the samples.
Pathogenicity	The pathogenicity for a selected variant. Default values are Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign.
Pathogenicity Status	Indicates the status for changing the pathogenicity for a selected variant. Default values are Pending and Confirmed.
Sample Status	The status for a sample. Default values are New, QC Passed, Reviewed, and Complete.
Panel Group	Separately tracks the pathogenicities of variants that might be found in multiple panels. For example, the pathogenicity of a selected variant is set to Deleterious for multiple cancer panels; however, the pathogenicity can be set to Benign for a cardiac panel. You can create a group for each panel, and then assign the appropriate panels to each group.
Coverage Region Status	The Coverage Regions tab (via the Run tab) 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 192 .
Sample Group	Used for a group of samples that are to be processed repeatedly over a period of time, for example, a group of control samples for which the coverage or variant frequency is to be tracked.
Run Status	The status for a run. Default values are New, QC Passed, Reviewed, and Complete.
Artifact Type	The artifact type for the sample variant, for example, a Sequencing artifact. A single default value, None, is supplied for artifact type.
Phenotype	The phenotype for a patient.

To add a value for an object

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

The Add/Edit Objects dialog box opens.

Figure 2-1: Add/Edit Objects dialog box



2. On the Object dropdown list, select the object for which you are adding a value.
If values have already been added for the selected object, or the selected object has default values, then these values are displayed in the Current Values pane.
3. In the Value field, enter the value for the object, and then click Add.



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

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

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



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



If you add a value for pathogenicity, you can also specify a highlighting color for variants of this type in all the variant panes and sub-tabs. See [“To specify the highlighting colors for variants of a pathogenicity”](#) on page 56.

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 50](#).

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

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

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

The value is highlighted in a blue rectangle.

4. Edit the value as needed.



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

5. Click OK.

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

To delete a value from an object



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

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

The Add/Edit Objects dialog box opens. See [Figure 2-1 on page 50](#).

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.

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Geneticist Assistant Setup and Maintenance

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

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

4. Click OK.

The Add/Edit Objects dialog box closes.

Specifying your Geneticist Assistant Settings

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

- Create your reference directory and specify its location. See [“To configure the reference directory” on page 54](#).
- Specify the global Quality Control (coverage) settings for your data. See [“To specify the global Quality Control settings” on page 55](#).

Optionally, you can also:

- Specify the colors for highlighting variants of a specific pathogenicity in all the variant panes and variant sub-tabs. See [“To specify the highlighting colors for variants of a pathogenicity” on page 56](#).
- Specify the same column display preferences for variants in all the variant panes and variant sub-tabs (with the exception of the Sample Group columns on the Sample Variant Data sub-tab for the Sample tab and the Variant pane on the Variant tab), and/or clear the preferred settings for all the data tabs. See [“To apply shared preferences or to clear all tab preferences” on page 58](#).
- 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 60](#).
- Specify your Alamut settings. See [“To specify Alamut settings” on page 61](#).
- Specify your HGMD settings. See [“To specify HGMD settings” on page 62](#).
- Specify the requirements for all user passwords for Geneticist Assistant. See [“To specify the Geneticist Assistant password requirements” on page 63](#).
- Specify your OMIM settings. See [“To specify the OMIM settings” on page 65](#).
- Configure your proxy server settings. See [“To configure proxy server settings” on page 66](#).
- Modify the SoftGenetics license server settings. See [“To modify the SoftGenetics license server settings” on page 66](#).

To configure the reference directory

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



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

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

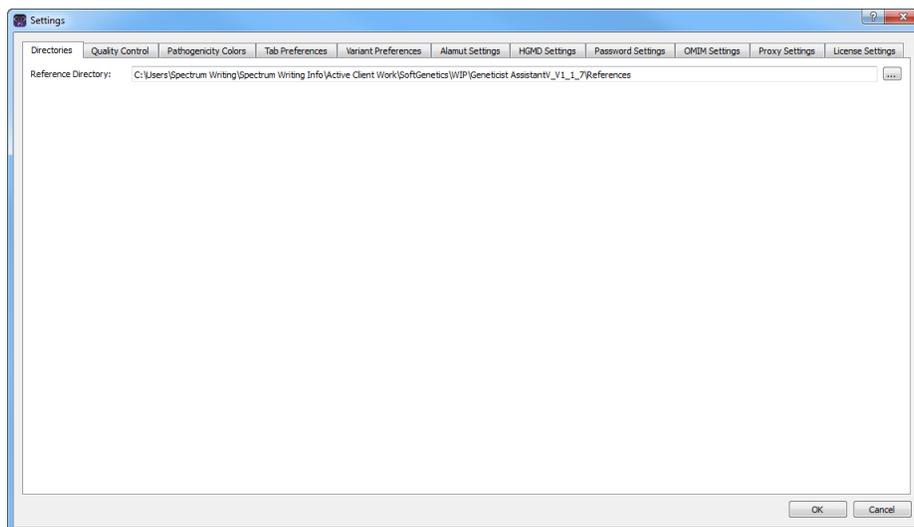


Although not required, SoftGenetics recommends that you create the reference directory on the same computer and drive on which you installed the Geneticist Assistant client.

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

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

Figure 2-2: Settings dialog box, Directories tab



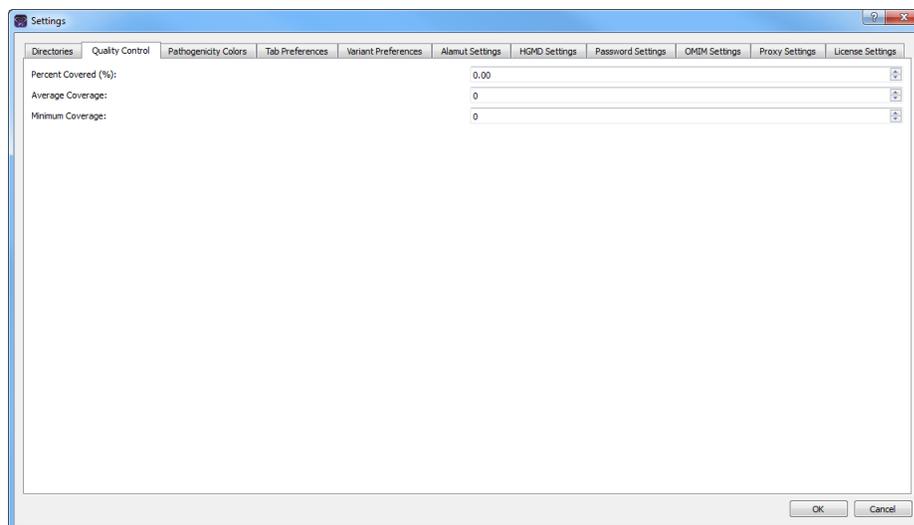
3. Click the Browse button  next to the Reference Directory field, and then browse to and select the Reference directory.
4. If you are done specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed.

To specify the global Quality Control settings

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

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

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



3. Specify the value for the Quality Control settings.
 - Minimum Coverage—The minimum number of reads that are required for a panel region.
 - Average Coverage—The average number of reads that are required for a panel region.
 - Percent Covered (%)—The minimum percentage of the panel region that has reads aligned to it.
4. If you are done specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed.

To specify the highlighting colors for variants of a pathogenicity

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

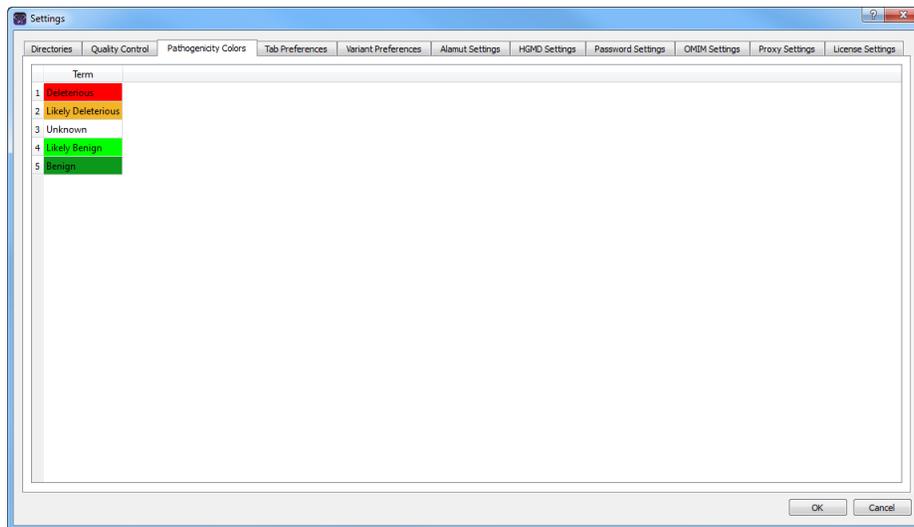
1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the Pathogenicity Colors tab.

The default highlighting colors for the five system-supplied pathogenicity values are displayed on the tab.



If you have added a custom pathogenicity value (see “[Managing Objects](#)” on page 49), then this value is also displayed, and if applicable, in its selected highlighting color.

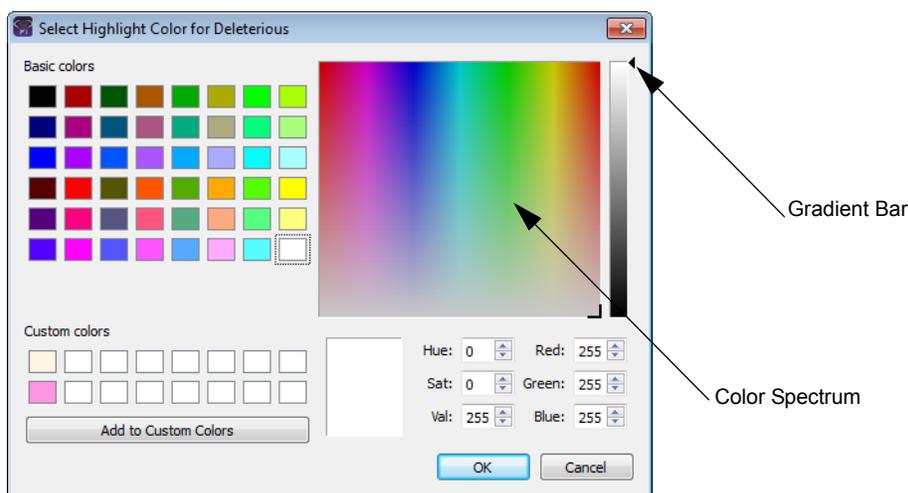
Figure 2-4: Settings dialog box, Default colors shown on the Pathogenicity Colors tab



3. Select a pathogenicity.

The Select Highlight Color dialog box opens. The pathogenicity that you selected is displayed in the title bar of the dialog box. See [Figure 2-5 on page 57](#).

Figure 2-5: Select Highlight Color dialog box



4. Continue to one of the following:
 - “To specify a basic highlighting color for a pathogenicity” on page 57.
 - “To specify a custom highlighting color for a pathogenicity” on page 58.



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

To specify a basic highlighting color for a pathogenicity

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

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

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

To specify a custom highlighting color for a pathogenicity

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

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

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

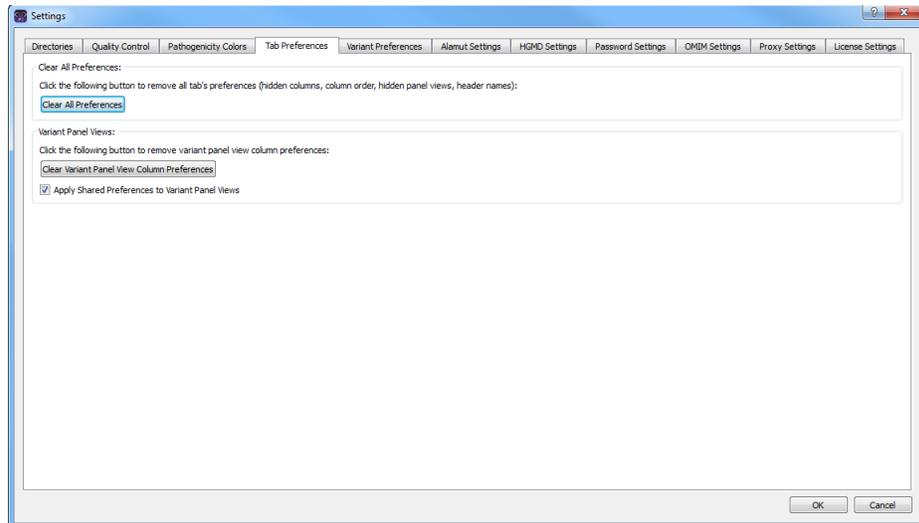
6. Continue selecting pathogenicities and specifying custom colors as needed.
7. If you are done specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs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

To apply shared preferences or to clear all tab preferences

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

1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.
2. Open the Tab Preferences tab. See [Figure 2-6 on page 59.](#)

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



3. Do one of the following:

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

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

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

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

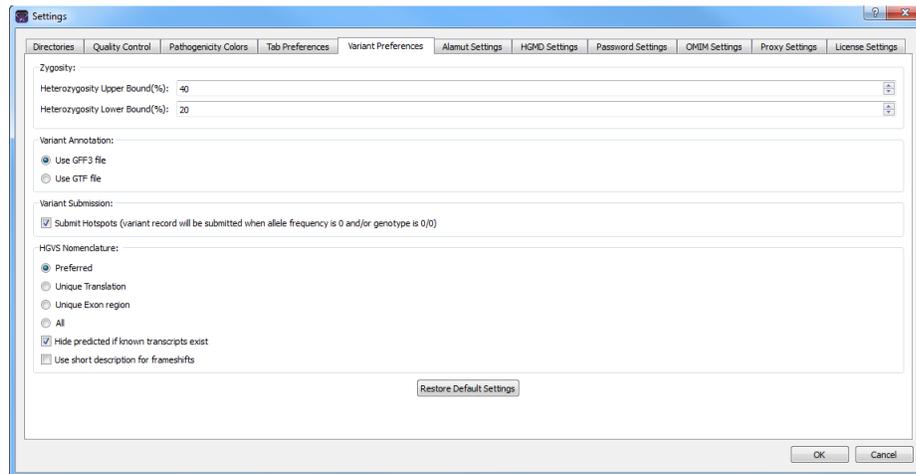
4. If you are done specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed.

To specify variant preferences

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

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

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



3. Set the variant preferences.

Option	Description
Zygoty	
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	
<ul style="list-style-type: none"> • Use GFF3 file 	<ul style="list-style-type: none"> • The default value. Use the annotation information in the reference files that SoftGenetics provides.
<ul style="list-style-type: none"> • Use GTF file 	<ul style="list-style-type: none"> • Use the annotation information in the reference files that ENSEMBL provides. <p>Note: If you select this option, then you must download the correct references files from ENSEMBL and place them in the Reference directory that is defined for your Geneticist Assistant installation. See “To configure the reference directory” on page 54.</p>

Option	Description
Variant Submission	
Submit Hotspots	Deselected by default. Any variants that are included in a VCF file that have an allele frequency value = 0 in the file <i>are</i> submitted to the Geneticist Assistant database.
HGVS Nomenclature - Determines which transcript is used for the HGVS nomenclature columns if a gene has multiple transcripts.	
<ul style="list-style-type: none"> • 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 90.
<ul style="list-style-type: none"> • Unique Translation 	Show multiple transcripts if the translation varies among the transcripts.
<ul style="list-style-type: none"> • Unique Exon region 	Show multiple transcripts if the exon regions vary among the transcripts.
<ul style="list-style-type: none"> • 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.

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 modify the information on any of the other tabs as needed.

To specify Alamut settings

To view variants that were imported into Geneticist Assistant in Alamut:

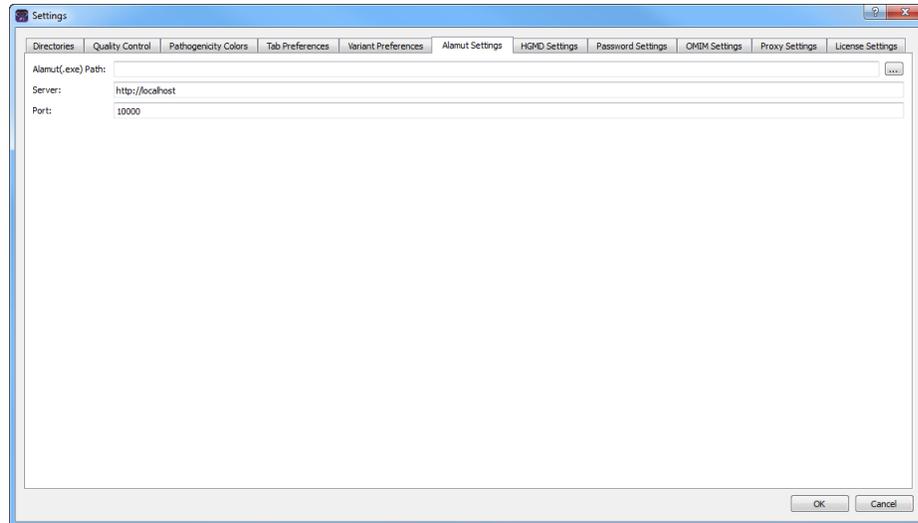
- HTTP-based access must be enabled in Alamut. To enable this in Alamut, select Tools > Options > Api, select Enable HTTP-based access, and then set the port to 10000.
- Geneticist Assistant must be able to connect to Alamut and *Alamut must be installed on the same computer as the Geneticist Assistant client.*
- Alamut must be open to jump to the variant position from Geneticist Assistant.

You specify the Alamut connection settings for Geneticist Assistant on the Alamut Settings tab.

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

2. Open the Alamut Settings tab.

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



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



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

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



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

6. 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 modify the information on any of the other tabs as needed.

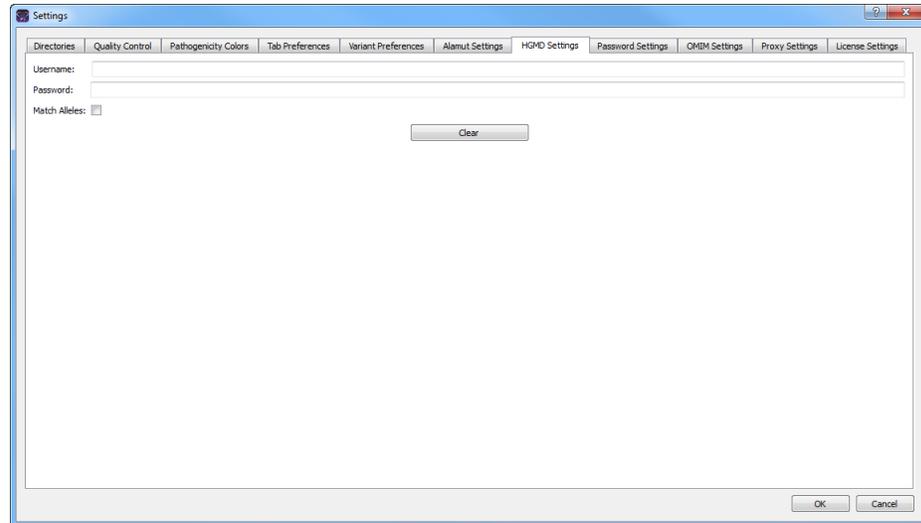
To specify HGMD settings

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

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

2. Open the HGMD Settings tab.

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



3. Enter your HGMD username and password.
4. Optionally, to show only the HGMD ID when the HGMD entry matches not only for the chromosome position, but also for the reference and alternate alleles, select Match Allele.
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 modify 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. By default, the only requirement is that the password cannot contain any spaces. You can, however, specify additional requirements that are applicable for all user passwords, including the Administrator password. You can specify the following requirements for your user passwords:

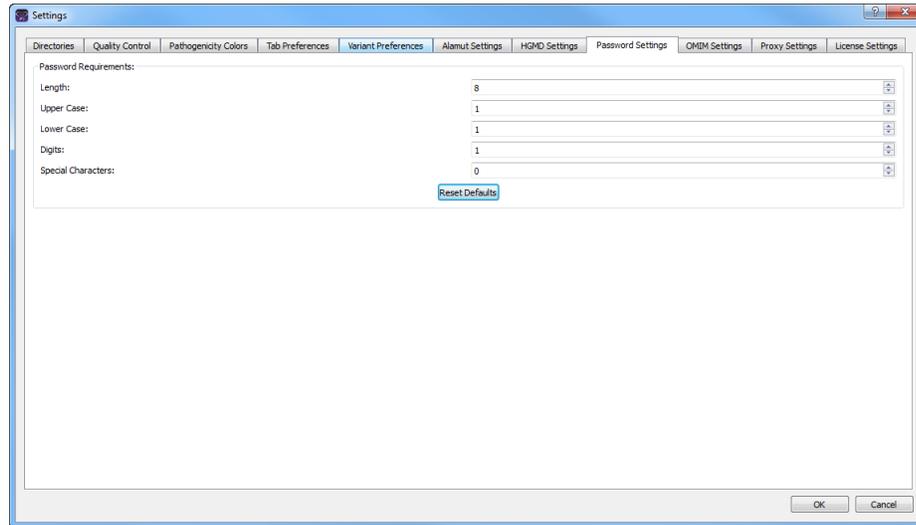
- Minimum length.
 - Minimum number of upper case characters.
 - Minimum number of lower case characters.
 - Minimum number of numeric characters.
 - Minimum number of special characters.
1. If you have not already done so, on the Geneticist Assistant main menu, click File > Settings to open the Settings dialog box.

Chapter 2
Geneticist Assistant Setup and Maintenance

2. Open the Password Settings tab.

A default value is provided for each password setting.

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



3. Specify the requirements for all user passwords for your Geneticist Assistant installation.



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

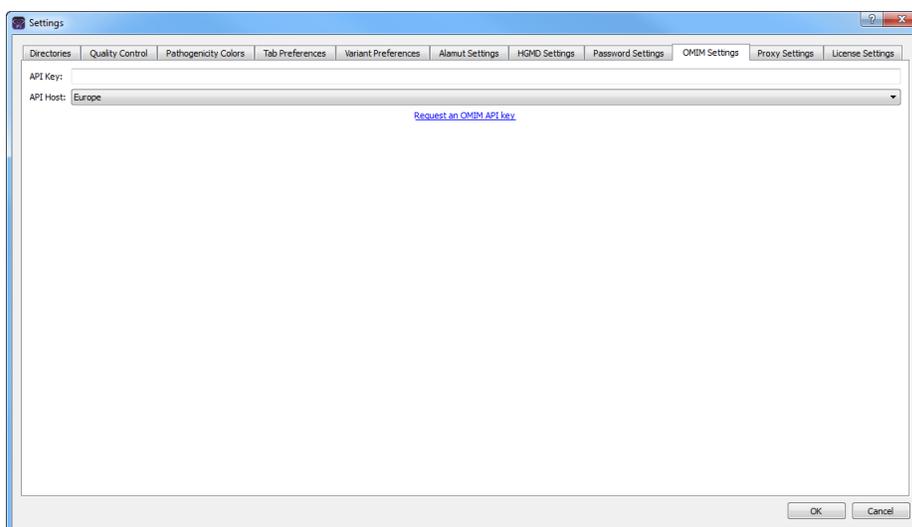
4. If you are done specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed.

To specify the OMIM settings

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

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

Figure 2-11: Settings dialog box, OMIM tab



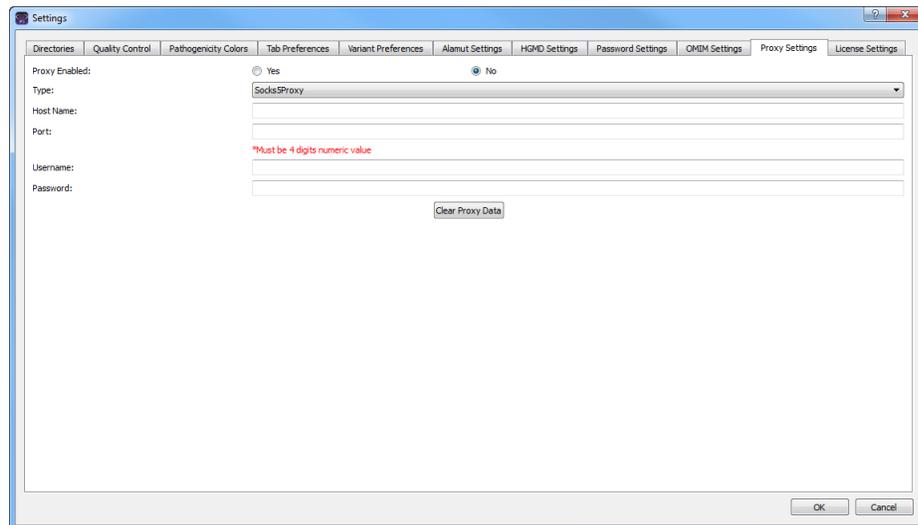
3. Click Request an OMIM API key.
You are directed to the API Access to OMIM webpage.
4. Complete the necessary information to obtain an OMIM API Access key.
5. Upon receipt of the key, enter the value in the API field.
6. Indicate the connectivity for the API Host (Europe or United States).
7. If you are done specifying your settings, then click OK to close the Settings dialog box and return to the Geneticist Assistant main window; otherwise, continue to add and/or modify the information on any of the other tabs as needed.

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

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



3. Configure your proxy server settings. If you select 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 modify the information on any of the other tabs as needed.

To modify the SoftGenetics license server settings

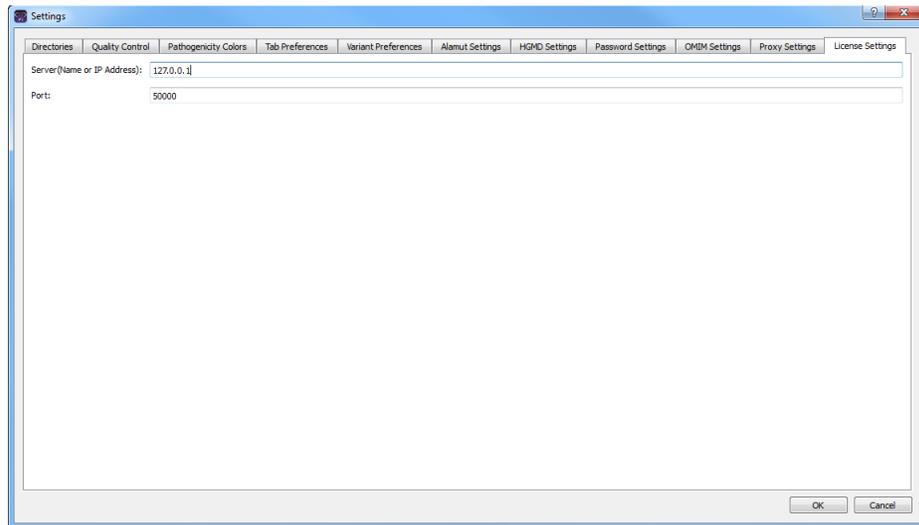
If you are running a purchased version of Geneticist Assistant, the *first* time that you run the program, you must configure Geneticist Assistant to connect to the SoftGenetics license server. (See [“To run a purchased version of Geneticist Assistant” on page 24.](#)) If you change the location of this license server, then you must modify the license server settings so that Geneticist Assistant can still connect to the server.

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

2. Open the License Settings tab.

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

Figure 2-13: Settings dialog box, License Settings tab



3. In the server name, 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.

4. Leave the Port value set to the default value of 50000.
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 modify the information on any of the other tabs as needed.

Backing up the Geneticist Assistant Database

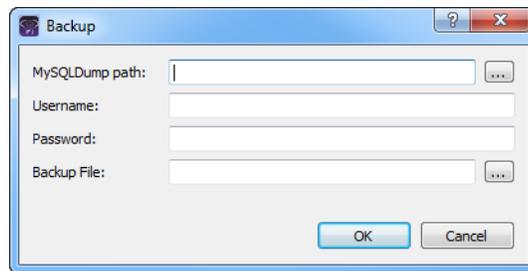
For data protection, you can manually **back up** the Geneticist Assistant database at any time of your choosing.

To back up the Geneticist Assistant database

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

The Backup dialog box opens.

Figure 2-14: Backup dialog box



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



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

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



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

5. Click OK.
After the Geneticist Assistant database is successfully backed up, a message opens indicating that the backup is finished.
6. Click OK to close the message.

The Geneticist Assistant main window remains open.

Chapter 3

Geneticist Assistant User Management

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

This chapter covers the following topics:

- [“Managing Users” on page 71.](#)
- [“Managing Groups” on page 77.](#)

Chapter 3
Geneticist Assistant User Management

Managing Users

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



Instead of assigning permissions to each user on an individual basis, optionally, you can assign users to a group and apply the same permissions to all the users in the group in a single step. See “[Managing Groups](#)” on page 77.

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.



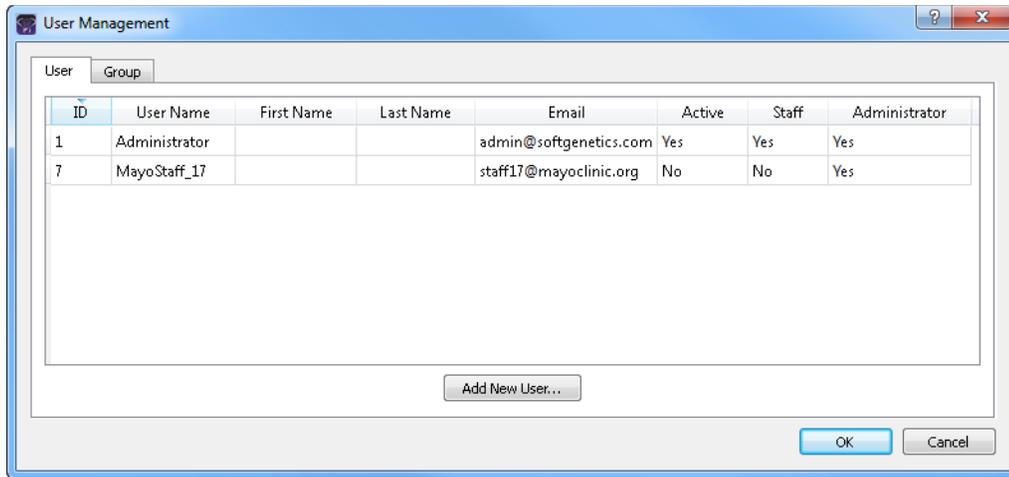
For a detailed description of the permission types that you can assign to a user, contact SoftGenetics.

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

The User Management dialog box opens. By default, the User tab is the open tab. The tab displays all the users who are currently configured for your Geneticist Assistant application, their status, and if applicable, their permissions level (Staff or Administrator). See [Figure 3-1 on page 72](#).

Chapter 3
Geneticist Assistant User Management

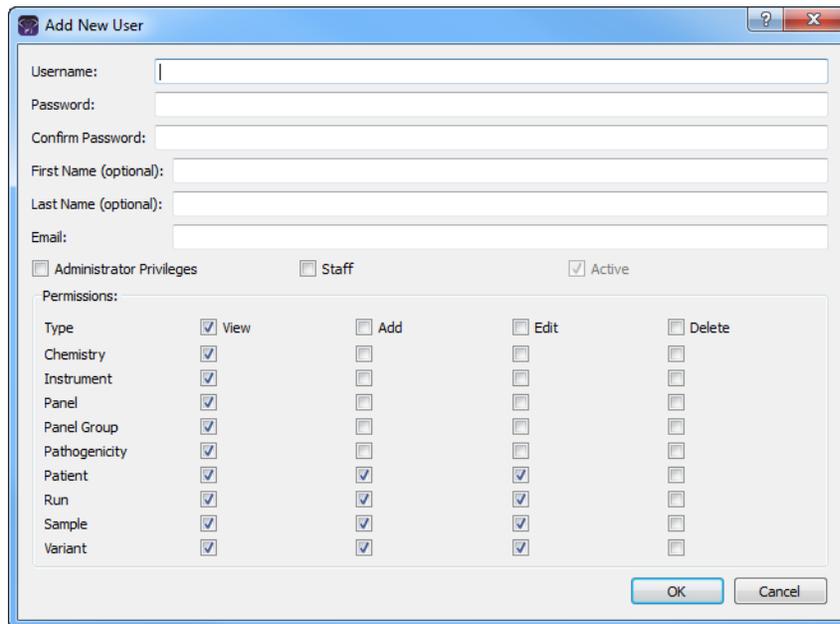
Figure 3-1: User Management dialog box, User tab



2. Click Add New User.

The Add New User dialog box opens.

Figure 3-2: Add New User dialog box



3. Add the information for the new user.

Field	Description
Username	Required. The login name for the user. Uppercase and lowercase are allowed. No special characters are allowed.
Password	Required. The login password for the user. The password cannot contain any spaces. If additional password requirements have been set, then the password must also adhere to these requirements. See "To specify the Geneticist Assistant password requirements" on page 63.
Confirm Password	Required. Re-enter the password exactly as you entered it in the Password field.
First Name (optional)	The first name for the user. Displayed only here in the application.
Last Name (optional)	The last name for the user. Displayed only here in the application.
Email	Required. The email address for the user.
<ul style="list-style-type: none"> • 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 75.

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



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

5. Click OK.

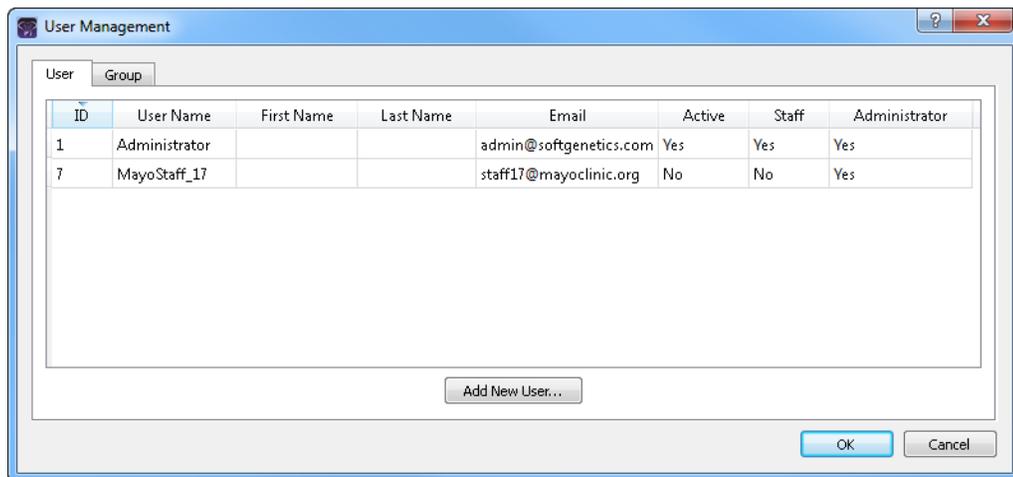
The User Management dialog box closes. You return to the Geneticist Assistant main window.

To delete a user

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

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

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



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

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

The user is deleted.

3. Click OK.

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

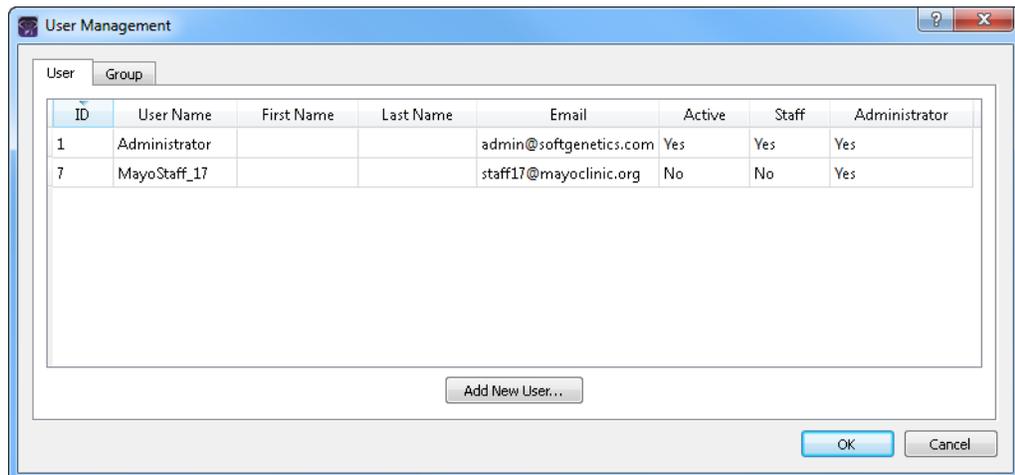
To edit a user

When you edit a user, you can modify the user password, the user first and last names, the user email address, the user status, the user level (Administrator and/or Staff) and the user permissions. You cannot edit a user name. To modify a user name, you must delete the existing user (see “[To delete a user](#)” on page 74), and then add the user with a new user name.

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

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

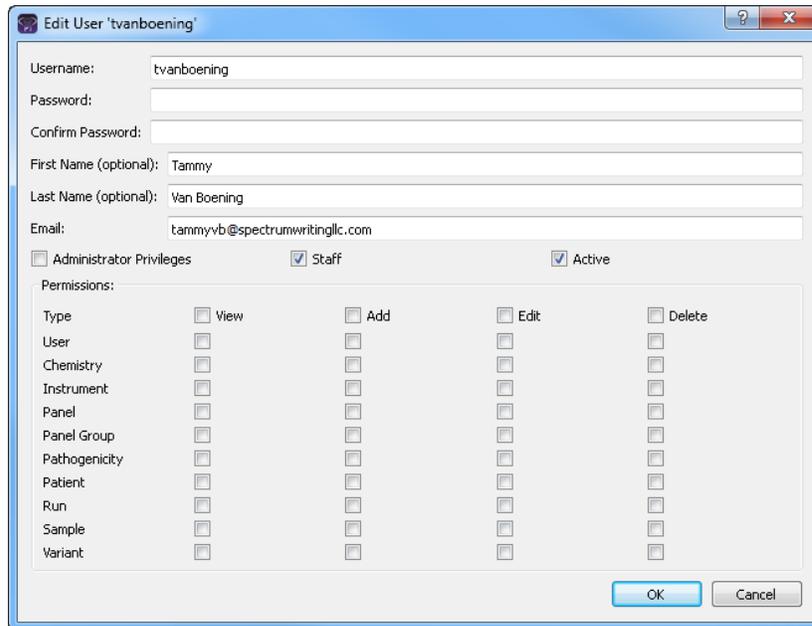
Figure 3-4: User Management dialog box, User tab



2. Do one of the following:
 - Double-click the user that you are deleting.
 - Right-click the user that you are editing, and on the context menu that opens, click Edit User.

The Edit User dialog box opens. The dialog displays all the current information for the user, including the currently assigned permissions. See [Figure 3-5 on page 76](#).

Figure 3-5: Edit User dialog box



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



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

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



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

5. Click OK.

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

6. Click OK.

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

Managing Groups

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

To add a new group

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

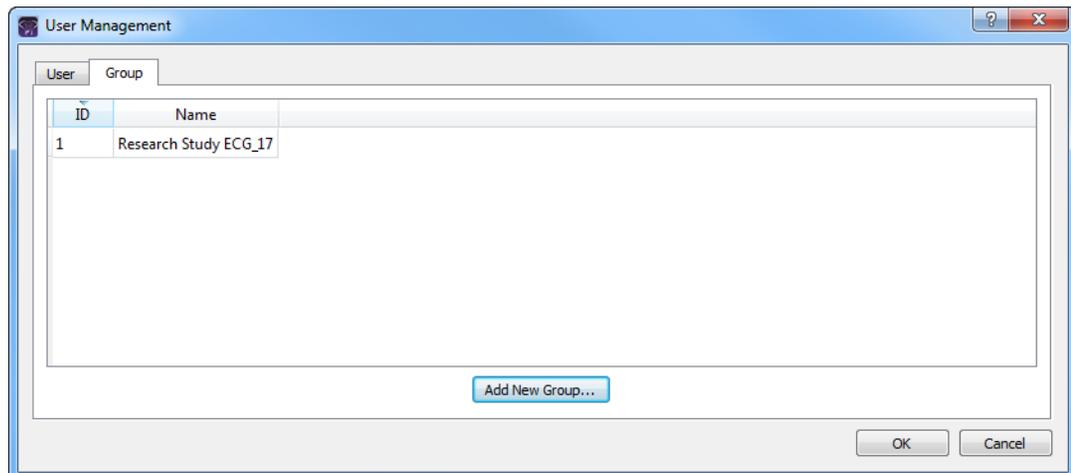
1. On the Geneticist Assistant main menu, click File > User Management.

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

2. Click the Group tab to open it.

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

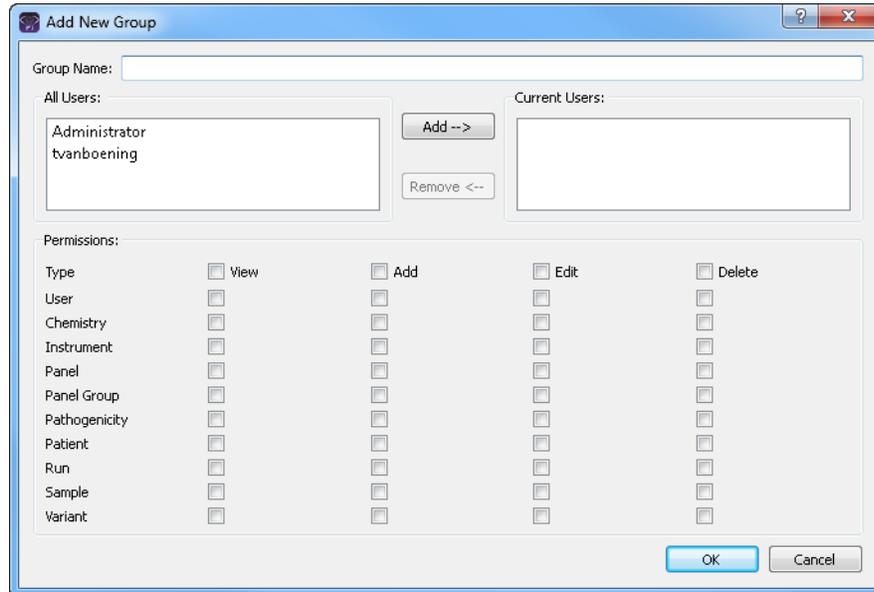
Figure 3-6: User Management dialog box, Group tab



3. Click Add New Group.

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

Figure 3-7: Add New Group dialog box



4. Enter the name for the new group.



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

5. In the All Users pane, select the users whom you are adding to the group (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.

8. Click OK.

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

To delete a group

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



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

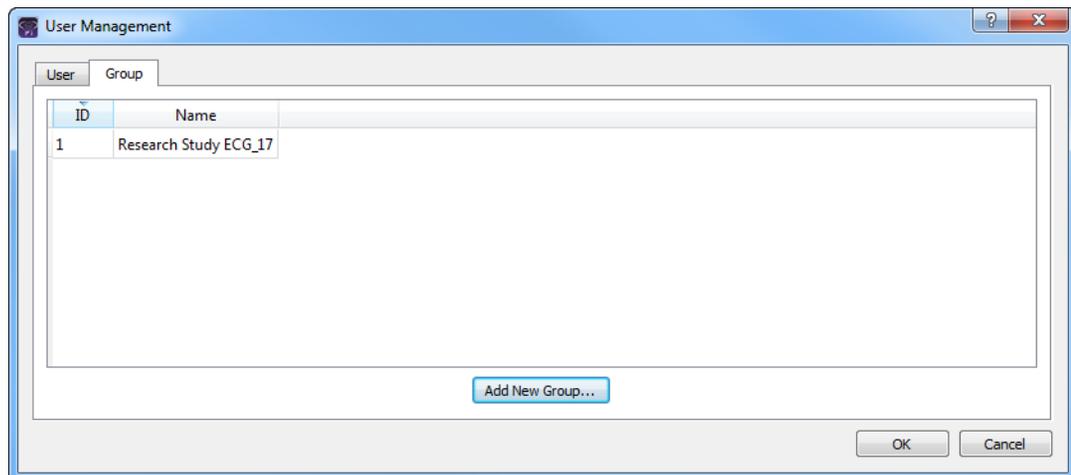
1. On the Geneticist Assistant main menu, click File > User Management.

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

2. Click the Group tab to open it.

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

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



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

The group is deleted.

4. Click OK.

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

To edit a group

When you edit a group, you can edit the users who belong to the group and the group permissions. You cannot edit the group name. To edit a group name, you must delete the existing group (see [“To delete a group” on page 79](#)), and then add the group with a new group name.

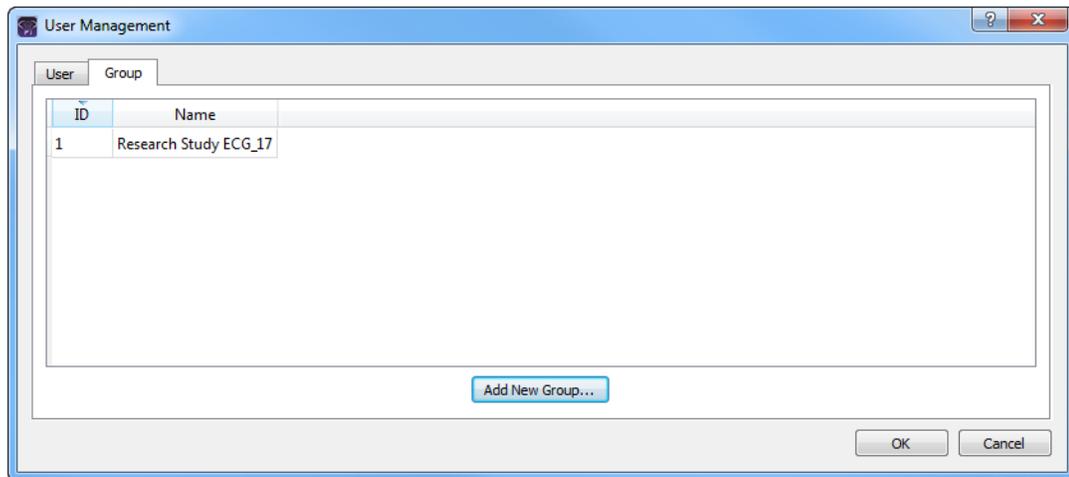
1. On the Geneticist Assistant main menu, click File > User Management.

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

2. Click the Group tab to open it.

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

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

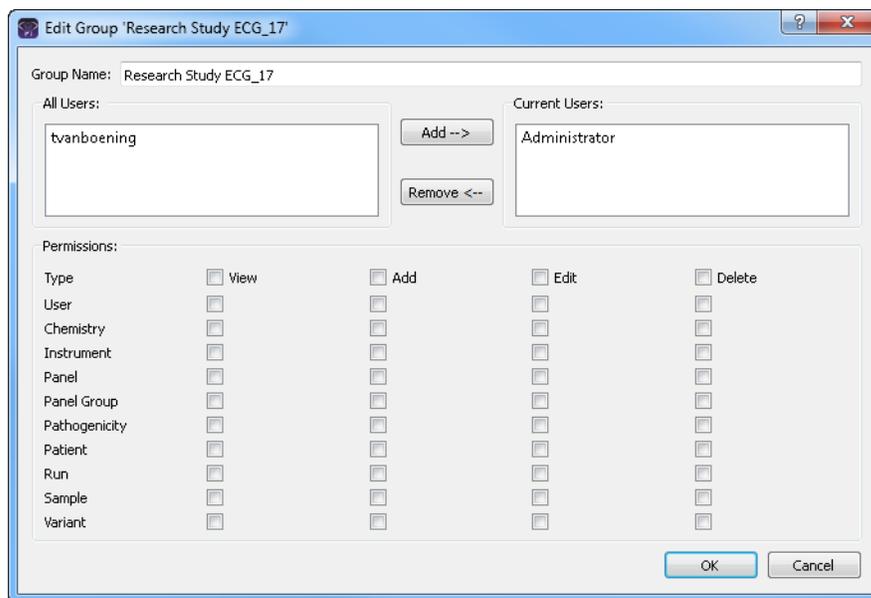


3. Do one of the following:

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

The Edit Group dialog box opens. The dialog displays all the current information for the group—the group name, the users who are currently assigned to the group, and the permissions for the group. See [Figure 3-10 on page 81](#).

Figure 3-10: Edit Group dialog box



4. To edit the users who belong to the group, do one or both of the following as needed:
 - To add users to the group—In the All Users pane, select the users whom you are adding to the group (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.

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Geneticist Assistant User Management

7. Click OK.

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

Chapter 4

Managing Panels and Subpanels

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

This chapter covers the following topics:

- [“Importing a Panel” on page 85.](#)
- [“Modifying a Panel” on page 95.](#)
- [“Deleting a Panel” on page 97.](#)
- [“Reviewing Panel Statistics” on page 98.](#)
- [“Adding a Subpanel” on page 99.](#)
- [“Deleting a Subpanel” on page 102.](#)

Chapter 4
Managing Panels and Subpanels

Importing a Panel

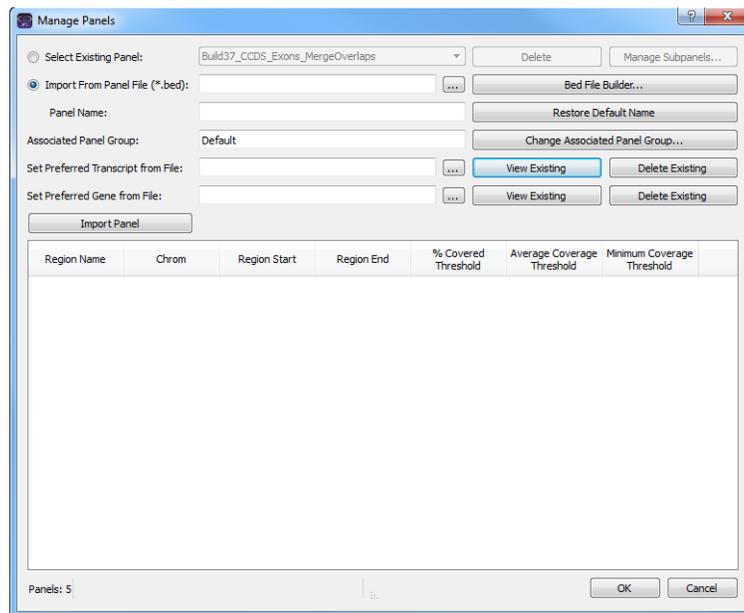
A *panel* is a collection of genomic regions that are targeted for a specific genomic test or study. Panels are added to Geneticist Assistant by **importing** a BED file that defines the targeted regions, known as *panel regions*.

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 93](#)), 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 279](#).

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.



For information about panel groups, see [“Managing Objects” on page 49](#).



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 55](#).

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.

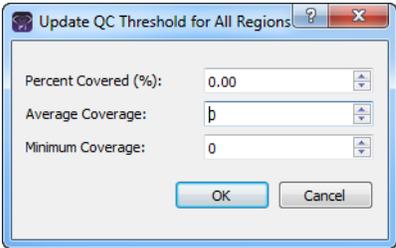
- If you are *not* done with the configuration of the newly added panel, then continue to any of the following as needed:
 - [“To manually set new coverage thresholds for panel regions” on page 87](#).
 - [“To create a QC Threshold file for panel regions” on page 88](#).
 - [“To set new coverage thresholds for panel regions with a QC Threshold file” on page 89](#).

- “To set or delete a preferred transcript list for a panel” on page 90.
- “To set or delete a preferred gene for a panel” on page 92.
- “To create a new BED file for a panel by adding or deleting regions” on page 93.

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. 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	<ol style="list-style-type: none"> 1. Click Update QC Threshold. The Update QC Threshold for All Regions dialog box opens. <i>Figure 4-2: Update QC Threshold for All Regions dialog box</i>  <ol style="list-style-type: none"> 2. For each coverage value that is to be edited, double-click it to select it, and then manually enter a new value, or click the Up/Down arrows to change the value accordingly. 3. Click OK.
To edit the values on a per region basis	<ol style="list-style-type: none"> 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.

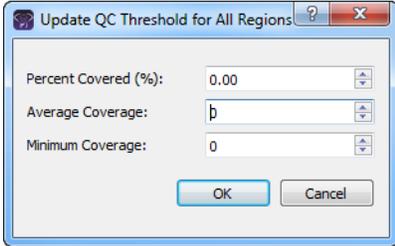
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Managing Panels and Subpanels

3. If you are done with the configuration of the newly added panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. (The edited coverage values are now saved for the panel in the Geneticist Assistant database.) Otherwise, continue any of the following as needed:
 - “To create a QC Threshold file for panel regions” below.
 - “To set or delete a preferred transcript list for a panel” on page 90.
 - “To set or delete a preferred gene for a panel” on page 92.
 - “To create a new BED file for a panel by adding or deleting regions” on page 93.

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	<ol style="list-style-type: none"> 1. Click Update QC Threshold. The Update QC Threshold for All Regions dialog box opens. <p><i>Figure 4-3: Update QC Threshold for All Regions dialog box</i></p>  <ol style="list-style-type: none"> 2. For each coverage value that is to be edited, double-click it to select it, and then manually enter a new value, or click the Up/Down arrows to change the value accordingly. 3. Click OK.

Action	Steps
To edit the values on a per region basis	<ol style="list-style-type: none"> 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. You can change one or both of these values. The file type is a text file, and you cannot change this.

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

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

5. If you are done with the configuration of the newly added or edited panel, then click OK to close the Manage Panels dialog box and return to the Geneticist Assistant main window. (The edited coverage values are now saved for the panel in the Geneticist Assistant database.) Otherwise, continue to one or both of the following as needed:
 - [“To set or delete a preferred transcript list for a panel”](#) on page 90.
 - [“To set or delete a preferred gene for a panel”](#) on page 92.
 - [“To create a new BED file for a panel by adding or deleting regions”](#) on page 93.

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

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

1. If you have just 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 88.

4. Click OK.

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

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

- [“To set or delete a preferred transcript list for a panel” on page 90.](#)
- [“To set or delete a preferred gene for a panel” on page 92.](#)
- [“To create a new BED file for a panel by adding or deleting regions” on page 93.](#)

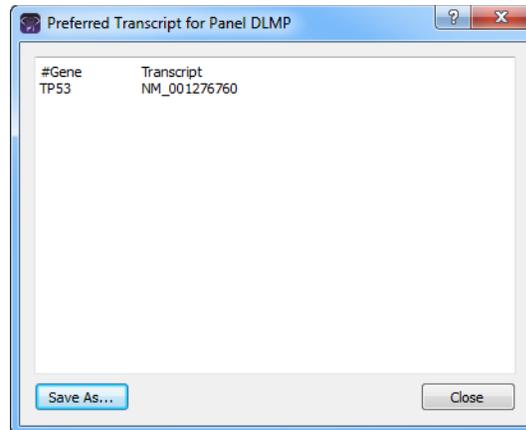
To set or delete a preferred transcript list for a panel

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. See [Figure 4-4 on page 91.](#)

Figure 4-4: 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 87.](#)
 - [“To create a QC Threshold file for panel regions” on page 88.](#)
 - [“To set new coverage thresholds for panel regions with a QC Threshold file” on page 89.](#)
 - [“To set or delete a preferred gene for a panel” on page 92.](#)
 - [“To create a new BED file for a panel by adding or deleting regions” on page 93.](#)

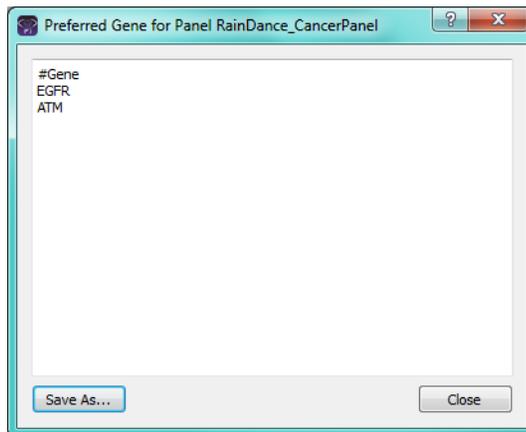
To set or delete a preferred gene for a panel

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 4-5: 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 87.](#)
 - [“To create a QC Threshold file for panel regions” on page 88.](#)
 - [“To set new coverage thresholds for panel regions with a QC Threshold file” on page 89.](#)
 - [“To set or delete a preferred transcript list for a panel” on page 90.](#)
 - [“To create a new BED file for a panel by adding or deleting regions” on page 93.](#)

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.

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Managing Panels and Subpanels

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

Action	Steps
To add regions from an existing BED file	<ol style="list-style-type: none"> 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	<ol style="list-style-type: none"> 1. Click the region that is to be deleted. (CTRL-click to select multiple regions.) 2. Right-click the selected regions, and on the context menu that opens, click Remove Selected Regions. <p>Note: Deleting regions from a panel does not delete the panel. To delete the panel itself, see “Deleting a Panel” on page 97.</p>

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 87](#).
- [“To create a QC Threshold file for panel regions” on page 88](#).
- [“To set new coverage thresholds for panel regions with a QC Threshold file” on page 89](#).
- [“To set or delete a preferred transcript list for a panel” on page 90](#).
- [“To set or delete a preferred gene for a panel” on page 92](#).

Modifying a Panel

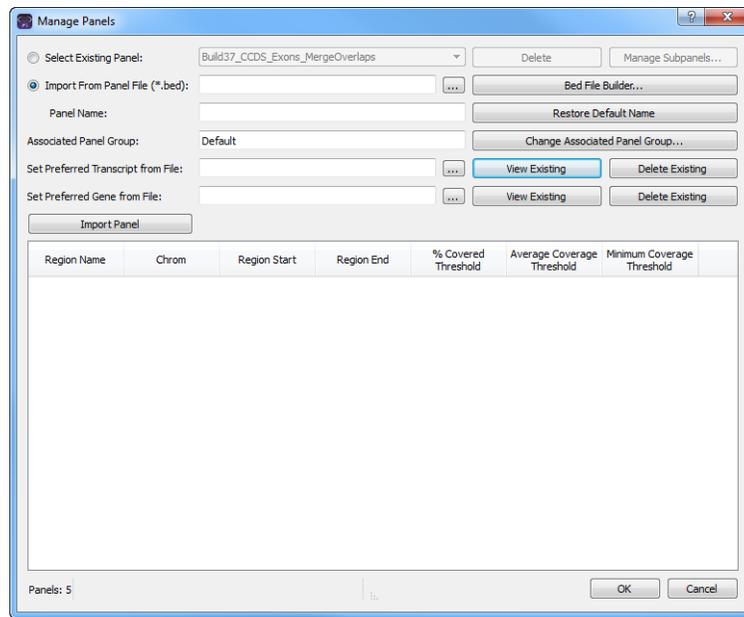
Managing panels includes [modifying](#) panels that will be used for studying your data in Geneticist Assistant.

To modify a panel

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

The Manage Panels dialog box opens.

Figure 4-6: *Manage Panels dialog box*



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

The panel regions are displayed on the Manage Panels dialog box.

4. Do any or all of the following as needed:
 - To change the associated panel group, click Change Associated Panel Group, and in the Panel Group Association dialog box, make the necessary change.
 - To manually set new coverage thresholds for the panel regions, see [“To manually set new coverage thresholds for panel regions”](#) on page 87.
 - 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 89.

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- 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 90](#).
- To set a preferred gene for the panel, see [“To set or delete a preferred gene for a panel” on page 92](#).
- 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 93](#).

Deleting a Panel

Managing panels includes [deleting](#) panels that are no longer needed in Geneticist Assistant.



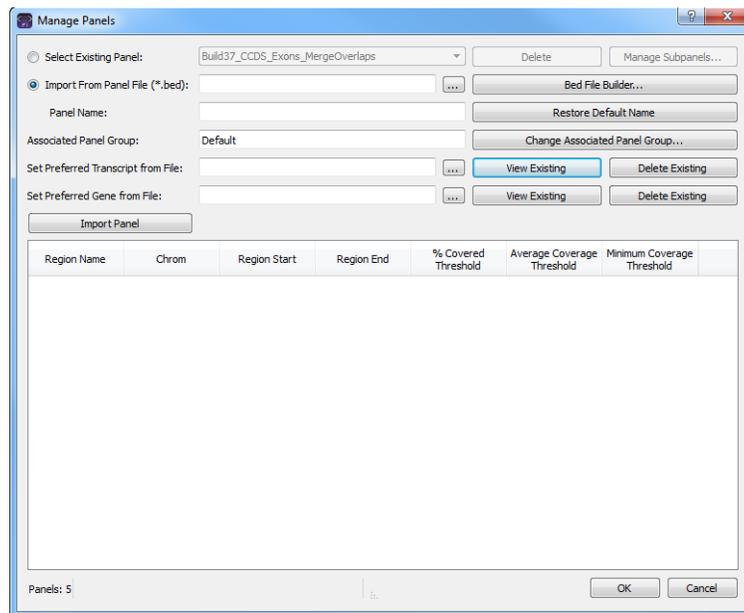
When you delete a panel, you delete the entire panel. To simply delete regions from a panel, not the entire panel, see “[To create a new BED file for a panel by adding or deleting regions](#)” on page 93.

To delete a panel

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

The Manage Panels dialog box opens.

Figure 4-7: *Manage Panels dialog box*



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

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

4. Click Yes.

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

5. Click OK.

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

Reviewing Panel Statistics

The Panel Statistics tab has options for [reviewing](#) specific information for a selected panel/panel group. By default, all samples for the panel are included. You can modify 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. 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 or the Run tab. For a complete description of this tab, see [“To calculate panel statistics \(Runs tab\)” on page 163](#) or [“To calculate panel statistics \(Run tab\)” on page 197](#).

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.

To add a subpanel

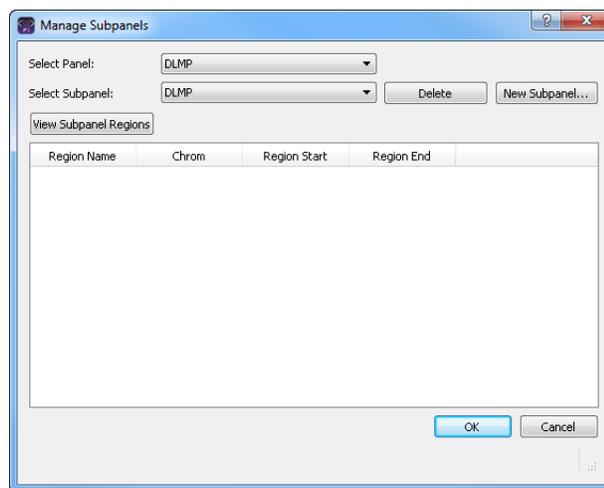
1. On the Geneticist Assistant main menu, click Panels > Manage Panels.

The Manage Panels dialog box opens. See [Figure 4-1 on page 85](#).

2. 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 4-8: *Manage Subpanels dialog box*

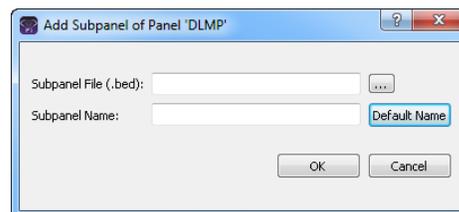


3. Select the panel for which you are adding the subpanel.

4. Click New Subpanel.

The Add New Subpanel dialog box opens.

Figure 4-9: *Add Subpanel dialog box*



5. Click the Browse button  that is next to the Subpanel File (.bed) field, 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 279.

6. Optionally, edit the name for the subpanel.



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

7. 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.

8. 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	<ol style="list-style-type: none"> 1. Click the Browse button  that is next to the Add Regions from BED File field. The Select a file to add to the panel dialog box opens. 2. Browse to and select the appropriate BED file, and then click Open to close the dialog box and return to the Manage Panels dialog box. 3. Click Add Regions.
To delete regions from the loaded BED file	<ol style="list-style-type: none"> 1. Click the region that is to be deleted. (CTRL-click to select multiple regions.) 2. Right-click the selected regions, and on the context menu that opens, click Remove Selected Regions. <p>Note: Deleting regions from a BED file does not delete the subpanel. To delete the subpanel itself, see "Deleting a Subpanel" on page 102.</p>

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.

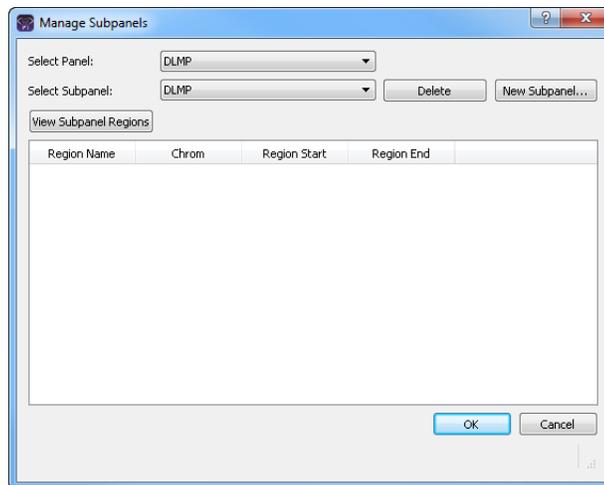


When you delete a subpanel, you delete the entire subpanel. To simply delete regions from a BED file, not the entire subpanel, see [“To create a new BED file for a subpanel by adding or deleting regions”](#) on page 100.

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 85](#).
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 4-10: Manage Subpanels dialog box

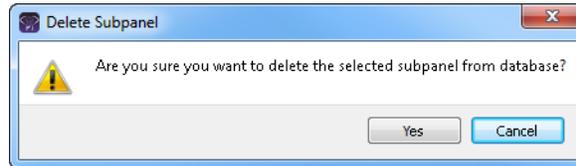


4. Select the subpanel that is to be deleted.

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.

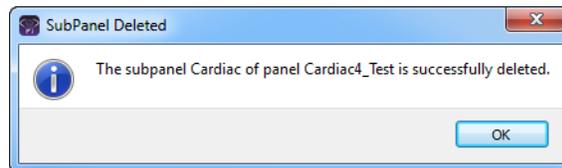
Figure 4-11: Delete Subpanel message



6. Click Yes.

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

Figure 4-12: 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.

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Managing Panels and Subpanels

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.

This chapter covers the following topics:

- [“Adding Patients” on page 107.](#)
- [“Accessing and Reviewing Patient Records” on page 112.](#)
- [“Adding a Run” on page 149.](#)



For information about managing your run settings (chemistries, instruments, and so on), see [“Managing Objects” on page 49.](#)

Chapter 5
Managing Patients and Runs

Adding Patients

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

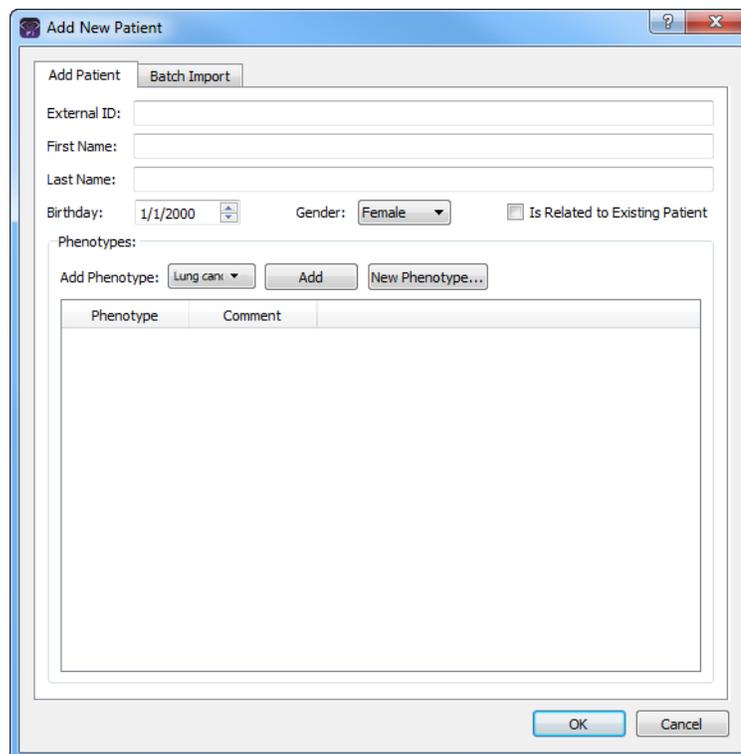
To add new patients one at a time

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

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

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

Figure 5-1: Add New Patient dialog box, Add Patient tab



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Managing Patients and Runs

2. Enter the information for the new patient.

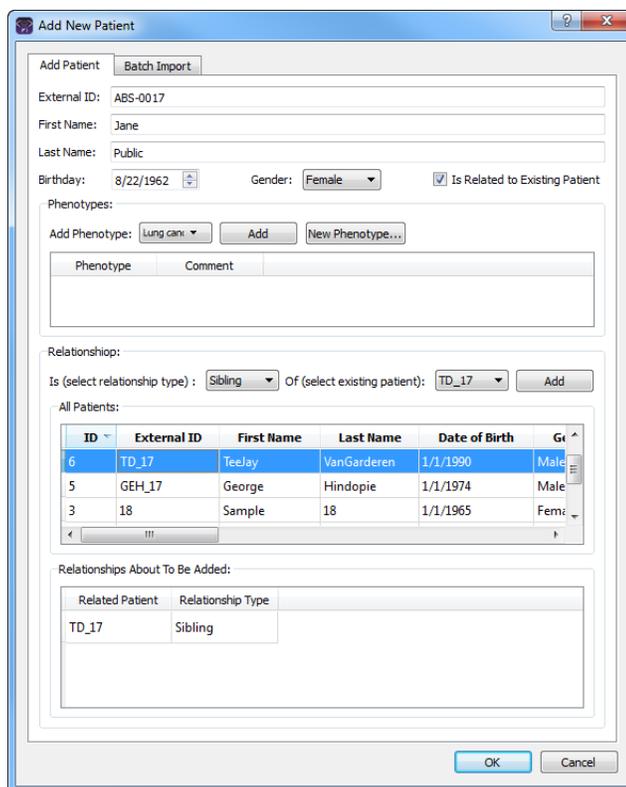
Option	Description
External ID	A unique ID for the patient.
First Name	The first name for the patient.
Last Name	The last name for the patient.
Birthday	The patient's birthday. You must select each value individually, and then you can manually enter a new value, or you can use the Up/Down arrows.
Gender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

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

- Select the relationship type (Sibling (the default value), Child, or Parent).
- Select the existing patient for this relationship. (Select the patient on the select existing patient list, or double-click the patient entry in the All Patients pane.)
- Click Add.

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

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



- 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.

- If applicable, to add a phenotype for the patient, select the phenotype on the Add Phenotype dropdown list, 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 and add it. You can also add phenotypes using the Manage Objects function. See “Managing Objects” on page 49.

- Optionally, to add a comment for the phenotype, do one of the following; otherwise, go to [Step 7](#).
 - Single click twice in the Comment field, 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.

- Click OK.

The Add New Patient dialog box closes.

To add new patients with the batch import process

You use a batch import process to add multiple patients in a single step to the Geneticist Assistant database. The batch import file must be either a comma-separated values (.csv) file, a tab-delimited text (.txt) file, or a tab-separated values (.tsv) file. The file must contain the following columns of information—the patient ID, the patient last name, the patient first name, the patient date of birth, the patient gender, the patient race, 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. [Figure 5-3](#) below shows the standard column order and the standard column names. If your import file does not follow either the standard column order or the standard column names, then you must manually indicate the order of the columns in your file.

Figure 5-3: Standard column order and names

	A	B	C	D	E	F	G	H
1	patient_ID	patient_last_name	patient_first_name	patient_dob	patient_gender	patient_race	patient_phenotype	patient_relationships
2	1320476	Doe	John	5/6/1913	Male	Caucasian		
3	1320683	Doe	Jane	3/2/1920	Female	Caucasian		BC-13-20476 - Father
4	1315487	Doe	Jill	6/10/1946	Female	Caucasian	phe1	BC-13-20476 - Mother
5	800402	Doe	Jill	6/10/1946	Female	Caucasian	phe1	BC-13-20683 - Sibling
6	800418	Doe	Jill	6/10/1946	Female	Caucasian	phe1	
7	800451	Doe	Jill	6/10/1946	Female	Caucasian	phe2	
8	800456	Doe	Jill	6/10/1946	Female	Caucasian	phe2	

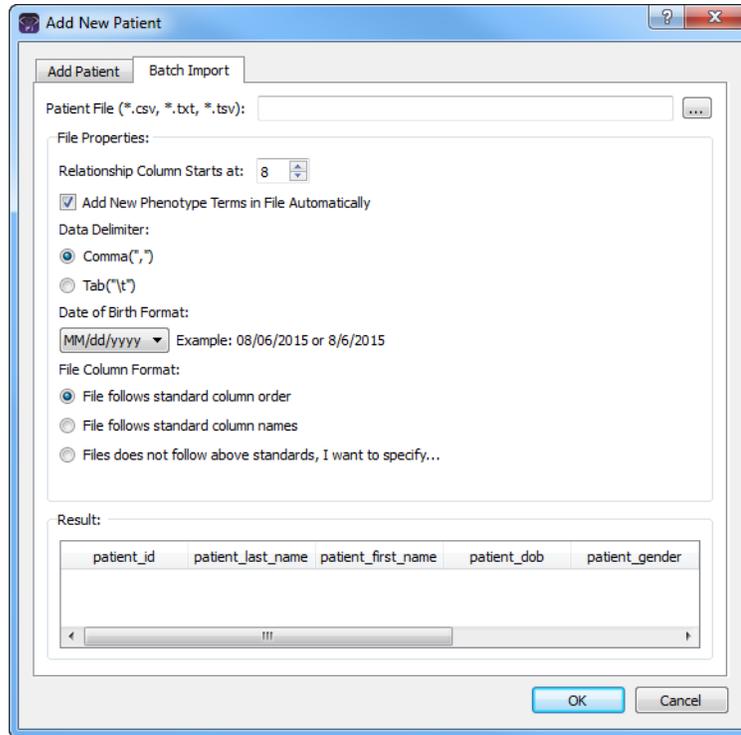
Chapter 5
Managing Patients and Runs

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

The Add New Patient dialog box opens. The Add Patient tab is the opened tab. See [Figure 5-1 on page 107](#).

2. Click the Batch Import tab to open it.

Figure 5-4: Add New Patient dialog box, Batch Import 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
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” on page 49), or the Add Phenotype option on the Add Patient tab (see “Patient tab, Patient sub-tab, Phenotype pane” on page 120), 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. <ul style="list-style-type: none"> • File follows standard column order. • File follows standard column names. • File does not follow above standards. I want to specify.

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



For information about rearranging columns, see “[To rearrange the order of data columns](#)” on page 42.

7. Click Check With File.
 - The file is loaded and the patient information is displayed in the Result pane. If your file format is correct, go to [Step 8](#).
 - If your file format is incorrect then, you must click Incorrect, Redo, and then make the necessary changes to the column headers. Click Check With File again to verify that your file has the correct format. Only after you verify that your file has the correct format can you continue to [Step 8](#).
8. Click OK.
The Add New Patient dialog box closes. The patients, and if applicable, any new phenotypes, are imported into your Geneticist Assistant database.

Accessing and Reviewing Patient Records

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

To review existing patient information

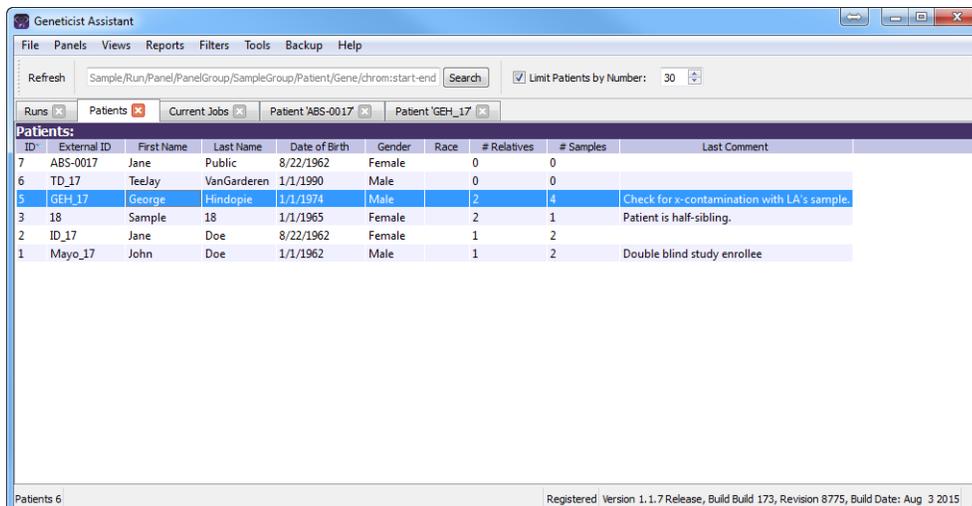
The Patients tab has a single pane, the Patients pane, for displaying information for existing patients. When you review the information for an existing patient, a variety of options are available for working with the patient, such as modifying the identifying information for a patient, and adding relationships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

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

The Patients tab opens.

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

Figure 5-5: Patients tab



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

Table 5-1: Patients tab context menu

Option	Description
Patient Detail	Opens the Patient tab. See Step 3 below. Note: You can also double-click a patient to open the Patient tab.
Add New Relationships	Opens the Add New Patient dialog box so that you can add a relationship to the selected patient. By default, the Is Related to Existing Patient option is selected and unavailable. Repeat Step 3 of “ To add new patients one at a time ” on page 107 as many times as needed to add all the necessary patient relationships to the patient.
Remove Patient's Relationships	Removes all the relationships that have been added for the patient.
Add All to Custom Report	Selects all the data that is displayed for the patients for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267 .
Delete Patient	Deletes the selected patient from the Geneticist Assistant database.
Add Comment	<p>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 124.</p> <p><i>Figure 5-6: Add Patient Comment dialog box</i></p> <div data-bbox="711 1367 1208 1814" data-label="Image"> </div> <p>Note: If multiple comments have been added for a patient, then the comment that was added last is displayed in the Last Comment field.</p>

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

Option	Description
Permissions	Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. If you are an Administrator user, then you can set or modify the permissions for a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See “To set or modify the Read/Write permissions for patient records” on page 115.
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 37. 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 58.
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 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.

- Do one of the following to open the Patient tab:
 - Right-click the patient record that you are reviewing, and on the context menu that opens, click Patient Detail.
 - Double-click the patient record that you are reviewing.

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

- [“Patient tab, Patient sub-tab” on page 116.](#)
- [“Patient tab, Samples sub-tab” on page 127.](#)
- [“Patient tab, Variants sub-tab” on page 139.](#)

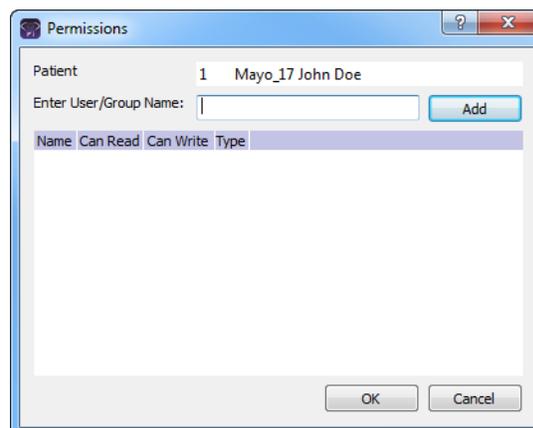
To set or modify the Read/Write permissions for patient records

1. If you have not already done so, open the Patients tab.
2. Select the patient or CTRL-click to select the multiple patients for which the permissions are being set.
3. Right-click any selected patient, and on the context menu that opens, click Permissions.

The Permissions dialog box opens.

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

Figure 5-7: Permissions dialog box (Patient tab)



4. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to [Step 5](#).



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.

5. 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.



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.

6. Select the appropriate 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. They cannot modify them.

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

8. Click OK.

A Permission Updated message opens.

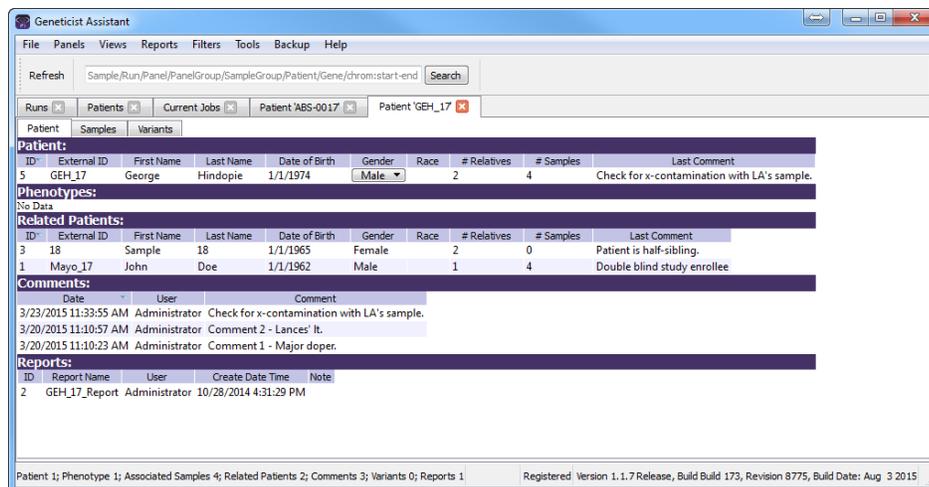
9. Click OK.

Both the message and the Permissions dialog box close. The specified permissions are set for the patient records.

Patient tab, Patient sub-tab

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

Figure 5-8: Patient tab, Patient sub-tab



The Patient sub-tab has five panes for displaying and working with patient information.

Pane	Available Options
Patient	<ul style="list-style-type: none"> Modifying the identifying information for a patient. Adding relationships to the patient. Adding comments for the patient. Removing relationships for a patient. See “Patient tab, Patient sub-tab, Patient pane” on page 117.
Phenotypes	Adding phenotypes for the patient. See “Patient tab, Patient sub-tab, Phenotype pane” on page 120.
Related Patients	<ul style="list-style-type: none"> Adding relationships to a related patient. Removing relationships for a related patient. See “Patient tab, Patient sub-tab, Related Patients pane” on page 122.
Comments	<ul style="list-style-type: none"> Adding comments for a patient. Deleting comments for a patient. See “Patient tab, Patient sub-tab, Comments pane” on page 124.
Reports	<ul style="list-style-type: none"> 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 126.

Patient tab, Patient sub-tab, Patient pane

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



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

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

Table 5-2: Patient tab, Patient sub-tab, Patient pane context menu

Option	Description
Add New Relationships	Opens the Add New Patient dialog box so that you can add a relationship to the selected patient. By default, the Is Related to Existing Patient option is selected and unavailable. Repeat Step 3 of “To add new patients one at a time” on page 107 as many times as needed to add all the necessary patient relationships to the patient.
Remove Patient’s Relationships	Removes all the relationships that have been added for the patient.
Add All to Custom Report	Selects all the data that is displayed for the patient for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267.

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

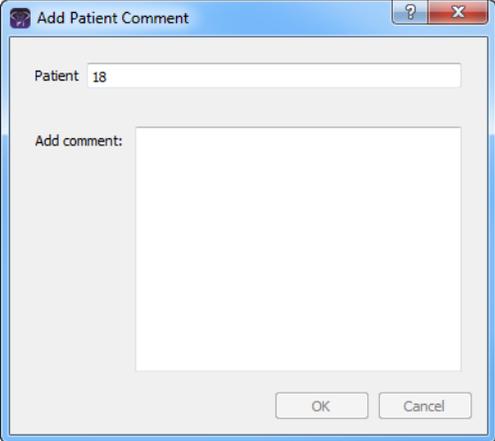
Option	Description
Add Comment	<p>Opens the Add Comment dialog box. You add a comment for the selected patient in the Add Comment field. The comment is displayed in the Last Comment field in the pane and in the Comments pane. See “Patient tab, Patient sub-tab, Comments pane” on page 124.</p> <p><i>Figure 5-9: Add Patient Comment dialog box</i></p>  <p>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.</p>
Permissions	<p>Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. If you are an Administrator user, then you can set or modify the permissions for a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See “To set or modify the Read/Write permissions for a patient record” below.</p>
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>
Compact	<p>Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.</p>

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

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 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.

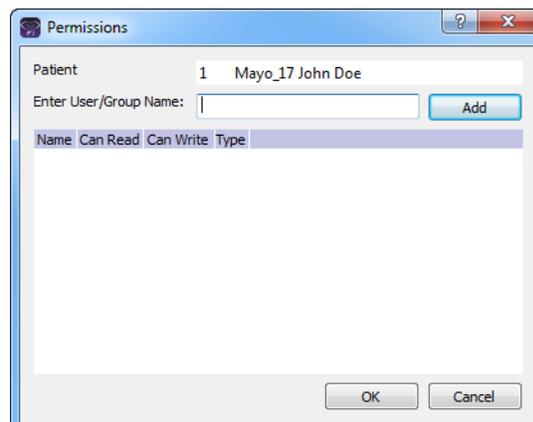
To set or modify the Read/Write permissions for a patient record

1. If you have not already done so, open the Patient sub-tab on the Patient tab.
2. Right-click the patient record, and on the context menu that opens, click Permissions.

The Permissions dialog box opens.

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

Figure 5-10: Permissions dialog box (Patient tab)



3. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to [Step 4](#).



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.



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 the appropriate 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 record. They cannot modify it.

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

7. Click OK.

A Permission Updated message opens.

8. Click OK.

Both the message and the Permissions dialog box close. The specified permissions are set for the patient record.

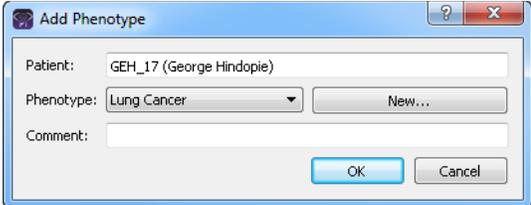
Patient tab, Patient sub-tab, Phenotype pane

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 5-3: 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 267.
Delete Phenotype	Deletes the selected phenotype for the patient.

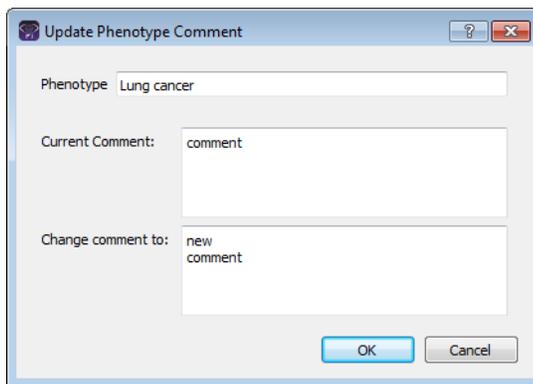
Table 5-3: Patient tab, Patient sub-tab, Phenotypes pane context menu (Continued)

Option	Description
<p>Add Phenotype</p>	<p>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.</p> <p><i>Figure 5-11: Add Phenotype dialog box</i></p>  <p>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” on page 49.</p>
<p>Load Columns</p>	
<p>Manually</p>	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>
<p>Compact</p>	<p>Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.</p>
<p><Saved Pane Configuration Name> - Saved by <User Name></p>	<p>Publicly available pane configurations. The name of the user who created a pane configuration is displayed next to the configuration name.</p>
<p>Load Filters</p>	
<p>Manually</p>	<p>Opens the Filters table for the Phenotypes pan. See “To filter the data that is displayed in a tab pane” on page 32.</p>
<p><Filter Template Name> - Saved by <User Name></p>	<p>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.</p>

You can single click twice in the Comment field in the Phenotypes pane to select the current comment in the field and edit it, or if the field is blank, enter a new comment. After you add or edit a comment, and then click out of the field, the Update Phenotype Comment dialog box opens. The dialog box displays the phenotype for which you are adding or editing the comment, the current (old) comment, and the new comment. You can make additional edits

to the new comment as needed, and then click OK to close the dialog box and save the new or modified comment for the phenotype.

Figure 5-12: Update Phenotype Comment dialog box



Patient tab, Patient sub-tab, Related Patients pane

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

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

Option	Description
Reload in Current Tab	Displays the information for the selected related patient in the currently opened Patient tab.
Open in New Tab	Displays the information for the selected related patient in a new Patient tab. The currently opened Patient tab remains open, but the focus is changed to a new Patient tab that displays the information for the related patient.
Add New Relationships	Opens the Add New Patient dialog box so that you can add a relationship to the selected related patient. By default, the Is Related to Existing Patient option is selected and unavailable. Repeat Step 3 of “ To add new patients one at a time ” on page 107 as many times as needed to add all the necessary patient relationships to the related patient.
Remove Patient's Relationships	Removes all the relationships that have been added for the patient.
Add All to Custom Report	Selects all the data that is displayed for all the related patients for inclusion in a custom report. See Chapter 10 , “ Geneticist Assistant Reports ,” on page 267 .

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

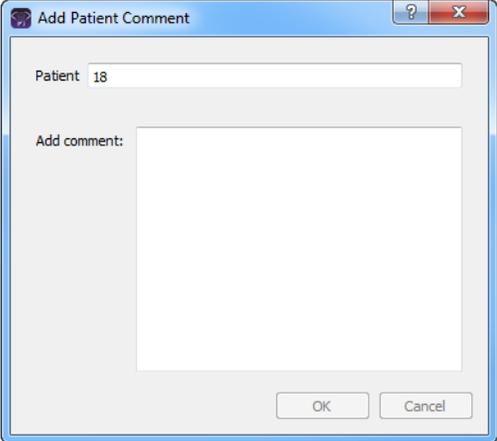
Option	Description
<p>Add Comment</p>	<p>Opens the Add Comment dialog box. You add a comment for the selected related patient in the Add Comment field. The comment is displayed in the Last Comment field in the pane.</p> <p><i>Figure 5-13: Add Comment dialog box</i></p>  <p>Note: If multiple comments 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.</p>
<p>Permissions</p>	<p>Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient record. If you are an Administrator user, then you can set or modify the permissions for a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See “To set or modify the Read/Write permissions for patient records” on page 115.</p>
<p>Load Columns</p>	
<p>Manually</p>	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>
<p>Compact</p>	<p>Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.</p>

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

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

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

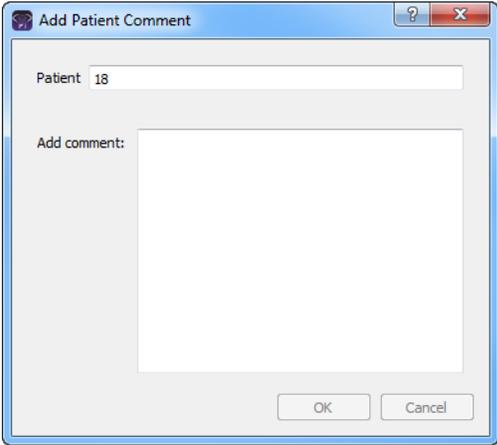
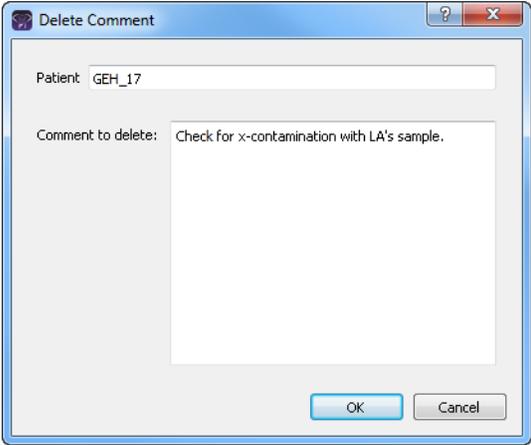
Option	Description
Add Comment	<p>Opens the Add Comment dialog box. You add a comment for the selected patient in the Add Comment field. The Comments are displayed in reverse chronological order in the pane.</p> <p><i>Figure 5-14: Add Comment dialog box</i></p> 

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

Option	Description
Delete Comment	<p>Opens the Delete Comment dialog box. Click OK to delete the comment for the patient and close the dialog box.</p> <p><i>Figure 5-15: Delete Comment dialog box</i></p> 
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>
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 5-6: Patient tab, Patient sub-tab, Reports pane context menu

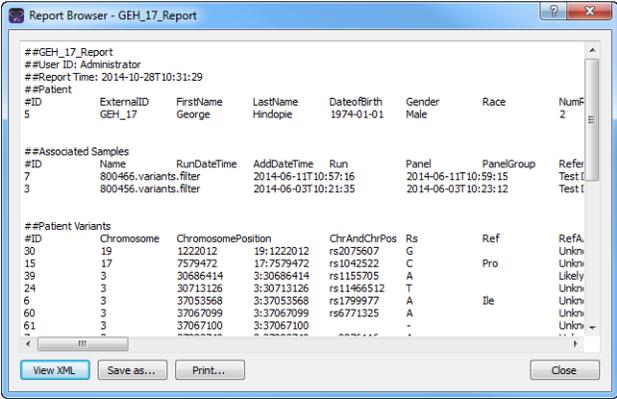
Option	Description
View Report	<p>Opens the Report Browser dialog box, which displays the report in plain text.</p> <p>Note: You can also double-click the report entry to open the Report Browser dialog box.</p> <p><i>Figure 5-16: Report Browser dialog box</i></p>  <p>The following options are available for the report:</p> <ul style="list-style-type: none"> • 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	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>

Table 5-6: 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.

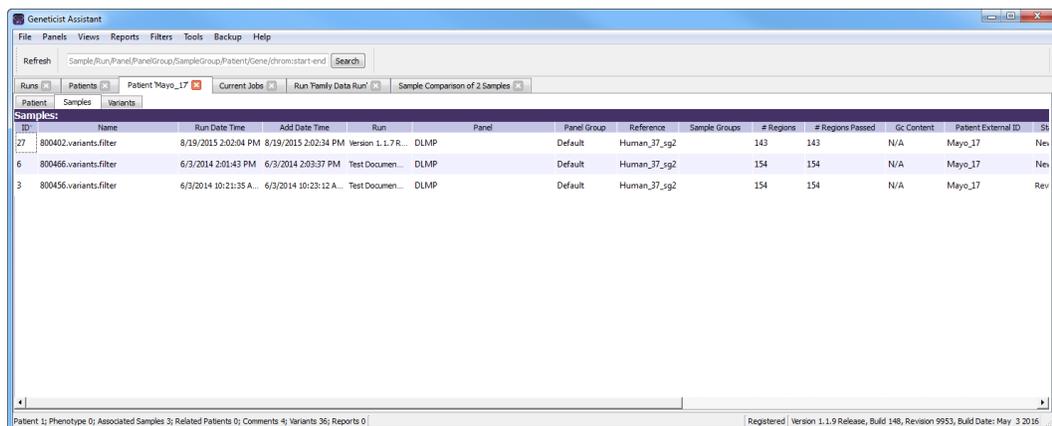


For detailed information about generating and saving both standard and custom Patient reports, see [Chapter 10, “Geneticist Assistant Reports,” on page 267.](#)

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 5-17: Patient tab, Samples sub-tab



You can do the following on the Samples sub-tab:

- View the information for an individual sample.
- View the coverage information for all regions in a sample panel.
- Delete the association between a selected sample and the patient and delete the sample from the Geneticist Assistant database.
- Generate a Batch Variants report for the selected sample.

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- Generate a Batch Coverage report for the selected sample.
- Add or edit a comment for a sample.
- Set sample permissions.

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

Option	Description
Sample Details	Opens the Sample tab, which contains detailed information about the associated sample, including the variant list. See Chapter 8, “Geneticist Assistant Data Review - Sample Tab,” on page 215. Note: You can also double-click the sample to open the Sample tab.
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 192.
Delete Sample	Deletes the association between the sample and the patient and removes the sample entry from the Samples pane.
Add All to Custom Report	Selects all the data that is displayed for all the associated samples in the run for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267.
Create Batch Variants Report	Generates the Batch Variants report for the selected associated sample or samples. (Press and hold the CTRL to select multiple samples.) See “To manage a Batch Variants report (Samples sub-tab on the Patient tab)” on page 130.
Create Batch Coverage Report	Generates the Batch Coverage report for the selected associated sample or samples. (Press and hold the CTRL to select multiple samples.) See “To generate a Batch Coverage report (Samples sub-tab on the Patient tab)” on page 136.

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

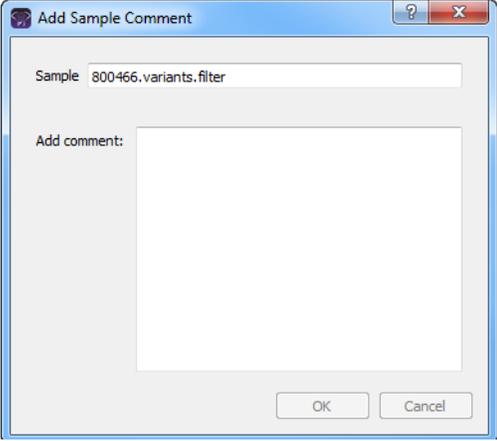
Option	Description
<p>Add Comment</p>	<p>Opens the Add Sample Comment dialog box. You add a comment for the associated sample in the Add Comment field. The comment is displayed in the Comments field in the Samples pane.</p> <p><i>Figure 5-18: Add Sample Comment dialog box</i></p>  <p>Note: If multiple comments have been added for a sample, then these comments are displayed in reverse chronological order in the Comments pane on the Sample Data sub-tab for the Sample tab. Only the last added comment is displayed in the Comments field in the Samples pane on the Samples data sub-tab on the Patient tab and in the Samples pane on the Run tab.</p>
<p>Permissions</p>	<p>Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the patient sample. If you are an Administrator user, then you can set or modify the permissions for a sample in a patient record that any user has added. If you are a standard user, then you can set or modify the permissions for samples in only those patient records that you have added to the Geneticist Assistant database, or for those patient records that other users have added and to which you have been granted Can Read and Can Write permissions. See "To set or modify the Read/Write permissions for a sample that is associated with a patient" on page 135.</p>
<p>Load Columns</p>	
<p>Manually</p>	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 37.</p> <p>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 58.</p>
<p>Compact</p>	<p>Displays the data columns on the sub-tab in a more condensed view. To toggle the display, click Compact as needed.</p>

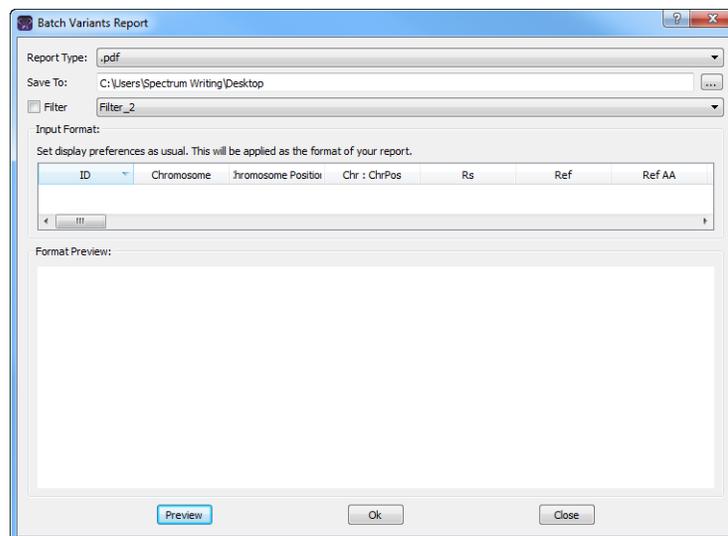
Table 5-7: Patient tab, Samples sub-tab, Samples pane context menu (Continued)

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 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.

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

A *Batch Variants report* is a report that includes information for variants in a selected sample, and allows you to save reports for multiple samples in a single step. A *Batch Variants report configuration* determines the data that is to be included in the report. You can 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 (PDF, HTML and so on), or the location in which the report is to be saved. Instead, you must specify the format and location in which to save the report every time you generate a report for a new or saved configuration.

Figure 5-19: Batch Variants Report dialog box



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

- [“To generate a Batch Variants report based on a new configuration” on page 131.](#)
- [“To generate a Batch Variants report based on a saved configuration” on page 132.](#)
- [“To modify a Batch Variants report configuration” on page 133.](#)

- [“To delete a Batch Variants Report configuration” on page 134.](#)

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 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.

1. If you have not already done so, open the Patient tab.
2. On the Samples sub-tab, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, on the Report Type dropdown list, select the report type. (The default value is PDF.)
5. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
 - Leave this location as-is, and then go to [Step 6](#).
 - 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.
6. Optionally, to filter the data that is to be displayed in the report, click 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.](#)

7. Optionally, to set the display preferences for the report, do any of the following as needed:
 - 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 Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.

10. Click OK.

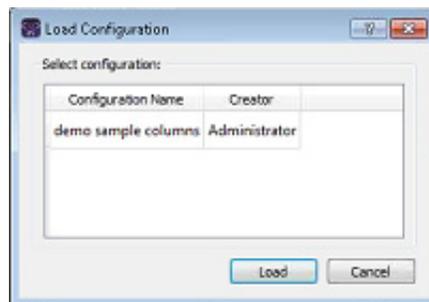
The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To generate a Batch Variants report based on a saved configuration

1. If you have not already done so, open the Patient tab.
2. On the Samples sub-tab, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, select the output format for the report.
5. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
 - Leave the location as-is, and then go to [Step 6](#).
 - 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.
6. Right-click on any heading in the Input Format pane, and then choose Select Columns. The Select Columns dialog box opens.
7. Click Load Configuration.

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

Figure 5-20: Load Configuration dialog box



8. Select a configuration, and then click Load.

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

9. Click Close (x) to close the Select Columns dialog box.
10. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.
11. Click OK.

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

12. Click OK.

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

To modify a Batch Variants report configuration

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

1. If you have not already done so, open the Patient tab.
2. On the Samples sub-tab, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.

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

4. 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. To modify the report configuration, see [“To create/modify a pane configuration” on page 39](#).

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

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

9. 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. If you have not already done so, open the Patient tab.
2. On the Samples sub-tab, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, right-click on any heading in the Input Format pane, and then choose Select Columns.
The Select Columns dialog box opens.
5. See [“To delete a pane configuration” on page 41](#).
6. Click Close (x) to close the Batch Variants Report dialog box.

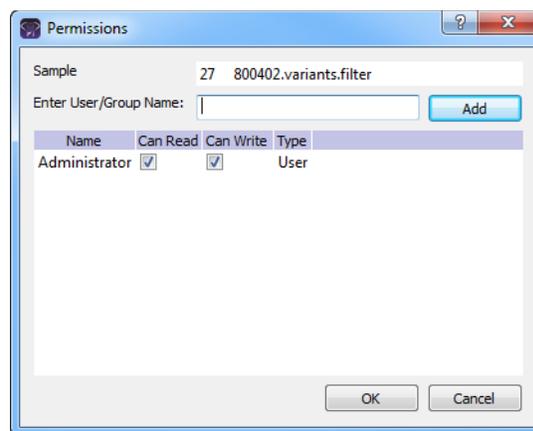
To set or modify the Read/Write permissions for a sample that is associated with a patient

1. If you have not already done so, open the Patient tab.
2. On the Samples sub-tab, select the sample or CTRL-click to select the multiple samples for which the permissions are being set.
3. Right-click any selected sample, and on the context menu that opens, click Permissions.

The Permissions dialog box opens.

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

Figure 5-21: Permissions dialog box (Samples sub-tab on the Patient tab)



4. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to [Step 5](#).



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.

5. 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.



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.

6. Select the appropriate 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 view the sample record. They cannot modify it.

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

To generate a Batch Coverage report (Samples sub-tab on the Patient tab)

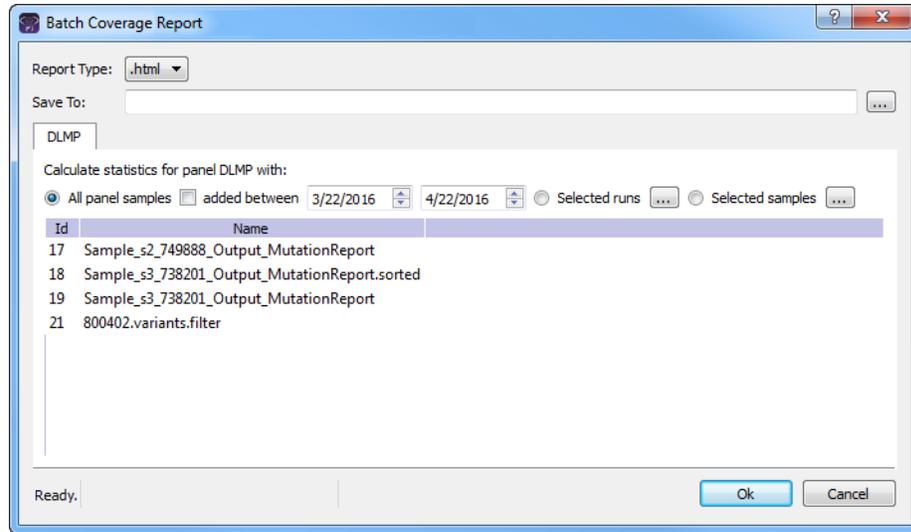
The Batch Coverage report generates a coverage report for each of the selected samples for the patient in a specific panel or panels and panel group. The report also includes coverage statistics for the panel that are calculated based on one of the following options:

- All 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.
 - Selected panel samples.
1. If you have not already done so, the Patient tab.
 2. On the Samples sub-tab, select the sample or CTRL-click to select the multiple samples for which the Batch Coverage report is to be generated.

- Right-click on any selected sample, and on the context menu that opens, click Export > Create Batch Coverage report.

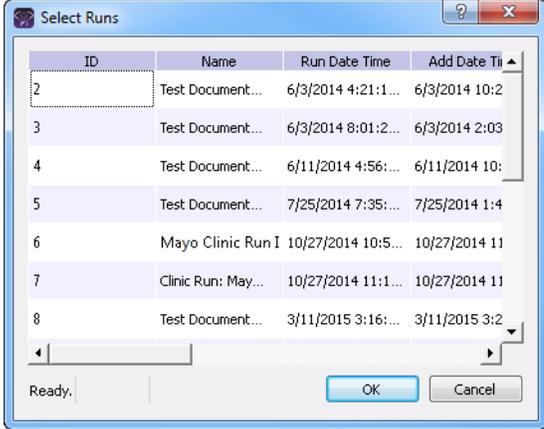
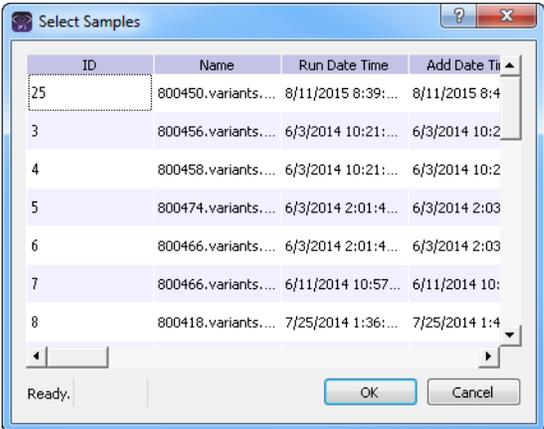
The Batch Coverage Report dialog box opens. All the selected samples for the run are displayed in the bottom pane of the dialog box. If multiple samples are selected with different panels, then a separate tab is displayed for each panel.

Figure 5-22: Batch Coverage Report dialog box (Runs tab)



- Leave the report type set to the default value of HTML, or select a different report type.
- 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.
- 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.

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: <ul style="list-style-type: none"> 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
<p>Selected runs</p>	<p>To select specific runs for calculating the panel coverage statistics, choose Selected runs, and then click the Browse button  that is next to Selected runs to open the Select Runs dialog box. By default, all the runs for the panel are displayed in the dialog box. Select the appropriate run, or CTRL-click to select multiple runs, and then click OK.</p> <p><i>Figure 5-23: Select Runs dialog box</i></p> 
<p>Selected samples</p>	<p>To select only specific samples for calculating the panel coverage statistics, choose Selected samples and then click the Browse button  that is next to Selected samples to open the Select Samples dialog box. By default, all the samples for the panel are displayed in the dialog box. Select the appropriate sample, or CTRL-click to select multiple samples, and then click OK.</p> <p><i>Figure 5-24: Select Samples dialog box</i></p> 



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 Runs tab.

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.

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. (See “Sample Variant Data sub-tab” on page 218.)

Figure 5-25: Patient tab, Variants sub-tab

Chromosome	Chromosome Position	Chr: ChrPos	Rs	Ref	Ref AA	Alt	Alt AA	Type	Coverage	Pathogenicity	Pathogenicity Status	Artifact Type	Gene	Gene Strand	Exon Number
3	37067094	3:37067094		TATATATT	-				46	Unassigned	None	None	MLH1	+	intron
17	63533599	17:63533599		T	Ile	C	Val	Synonymo...	563	Unassigned	None	None	AXIN2	-	6
17	63532528	17:63532528	rs138287857	G	Ala	A	Val	missense	288	Unassigned	None	None	AXIN2	-	8
3	37067100	3:37067100				T			38	Unassigned	None	None	MLH1	+	intron
3	37067099	3:37067099	rs8771325	A		T			38	Unassigned	None	None	MLH1	+	intron
2	48026757	2:48026757	rs267608078	AG	Glu	-	GlyfsTer16	frame-shift	659	Pathogenic	Pathogenic	None	MSH6	+	4
2	48032875	2:48032875	rs2324731	CTAT					205	Unassigned	Pending	None	MSH6	+	intron
2	48033891	2:48033891	rs267608137	T					311	Likely Benign	Likely Benign	None	MSH6	+	intron
22	29130458	22:29130458	rs1805128	T		C			99	Unassigned	None	None	CHEK2	-	5' UTR
2	47693941	2:47693941		C	Asn	-	ThrfsTer1	frame-shift	370	Likely Deleterious	Likely Deleterious	None	MSH2	+	11
18	48584856	18:48584856	rs386387676	-		TT			328	Unassigned	None	None	SMAD4	+	intron
10	89720634	10:89720634		T					241	Benign	Benign	None	PTEN	+	intron

You can specify how the information about the variants is to be displayed on the Variants sub-tab. See “To specify variant preferences” on page 60. You can also do the following for an individual variant that is displayed on the Variants sub-tab:

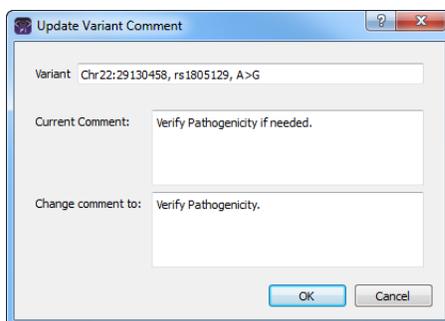
- Select a different gene for a variant: If the variant is located where there are overlapping genes, then you can select a different gene for the variant on the Gene dropdown list. If you select a different gene, then the gene that is used on the current tab for the current Geneticist Assistant session 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 92.
- 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*, 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 90](#).

- 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 Update Variant Comment dialog box opens.

Figure 5-26: Update Variant Comment dialog box



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



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

- Edit the type field, zygosity field, and/or the HGVS information (Genomic, Coding, and/or Protein) for a variant: You can click once in the Type field, the Zygosity field, the HGVS Genomic field, the HGVS Coding field, the HGVS Protein field, and the Read Balance field to select the current value in the field and edit it, or if the field is blank, enter a new value.



If you make a change to any of these values for the variant, then these changes are recorded for the variant on the Variant Changes pane on the Variant tab. See “Variant Changes pane” on page 253. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in the Geneticist Assistant application.

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

Table 5-8: 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 247. Note: You can also double-click the variant to open the Variant tab.
Show Variants Filtered by Panel	Displays all the variants in all the associated samples for the selected patient filtered by a selected panel. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected patient. <i>Figure 5-27: Select Panel to Filter Variants dialog box</i>
Show Samples with Selected Variants	Opens the Samples with variants pane, which displays a list of all the samples in your Geneticist Assistant database that contain the selected variant. 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 184.

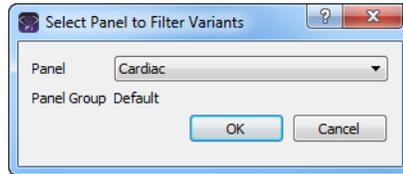


Table 5-8: Patient tab, Variants sub-tab context menu (Continued)

Option	Description
Edit Variant - Contains all the sub-menu options for editing and deleting a variant.	
Add Variant Comment	<p data-bbox="451 415 1365 646"> 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. </p> <p data-bbox="451 659 1015 690"> <i>Figure 5-28: Add Variant Comment dialog box</i> </p> <div data-bbox="662 705 1159 1150" data-label="Image"> </div> <p data-bbox="451 1178 1365 1320"> 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. </p>

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

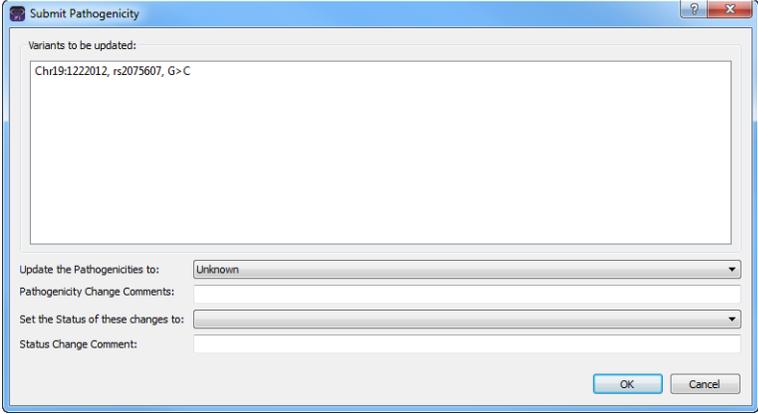
Option	Description
<p>Update Pathogenicity</p>	<p>Opens the Submit Pathogenicity dialog box. You can update the pathogenicity and/or pathogenicity status for the selected variant in this dialog box. You can also add a comment for the variant. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be. After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant.</p> <p><i>Figure 5-29: Submit Pathogenicity dialog box</i></p>  <p>Note: You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box. You also CTRL-click to select multiple variants for updating the pathogenicities and the pathogenicity statuses at the same time.</p> <p>Note: You can also update the Pathogenicity and the Pathogenicity Status for a variant on the Sample Variant Data sub-tab on the Sample tab.</p> <p>The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant:</p> <ul style="list-style-type: none"> • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See “Managing Objects” on page 49. • Optionally, you can add supporting information for this pathogenicity change. See “To add supporting information for a variant” on page 257. • You always have the option of updating just the pathogenicity status for a variant at a later date. See “Update Pathogenicity Status” below.

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

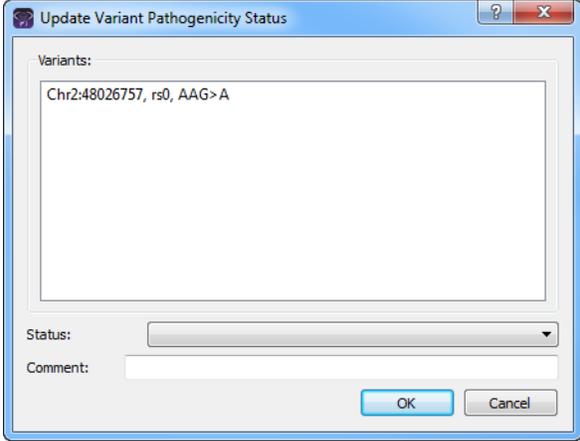
Option	Description
Update Pathogenicity Status	<p>Opens the Update Variant Pathogenicity Status dialog box. You can update the pathogenicity status for the selected variant in this dialog box. You can also add a comment for the variant.</p> <p><i>Figure 5-30: Update Variant Pathogenicity Status dialog box</i></p>  <p>The following information is relevant to updating the pathogenicity status for a variant:</p> <ul style="list-style-type: none"> • You can also double-click in the Pathogenicity Status column to open the Update Variant Pathogenicity Status dialog box. • You can CTRL-click to select multiple variants for updating the pathogenicity statuses at the same time. • You can also update the Pathogenicity Status for a variant on the Sample Variant Data sub-tab on the Sample tab. • By default, only two statuses, Pending or Confirmed, are available. You have the option of adding custom pathogenicity statuses. See “Managing Objects” on page 49.

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

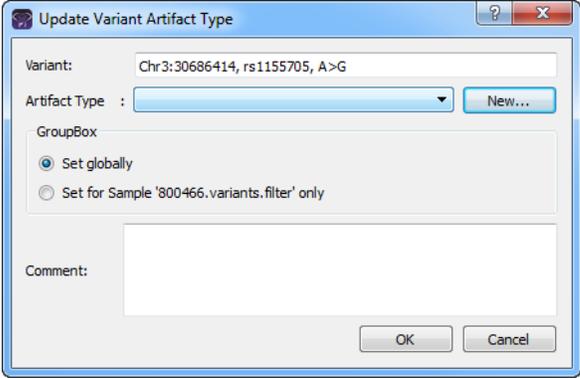
Option	Description
Update Artifact Type	<p>Opens the Update Artifact Type dialog box. You specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type.</p> <p><i>Figure 5-31: Update Variant Artifact Type dialog box</i></p>  <p>When you specify the artifact type, you can set the artifact type as the following:</p> <ul style="list-style-type: none"> • 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. <p>You also have the option of adding comments for the variant artifact type.</p> <p>The following information is relevant to updating the artifact type for a variant:</p> <ul style="list-style-type: none"> • You can CTRL-click to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant in the Variants pane, on the Variants sub-tab on the Panel Statistics tab from the Runs tab or the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See “Managing Objects” on page 49.

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

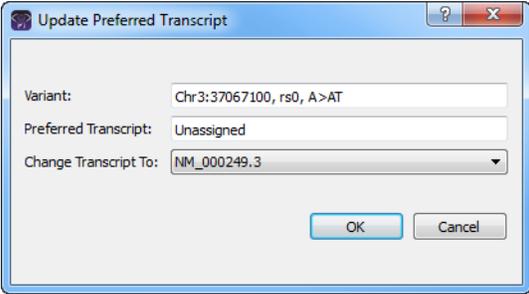
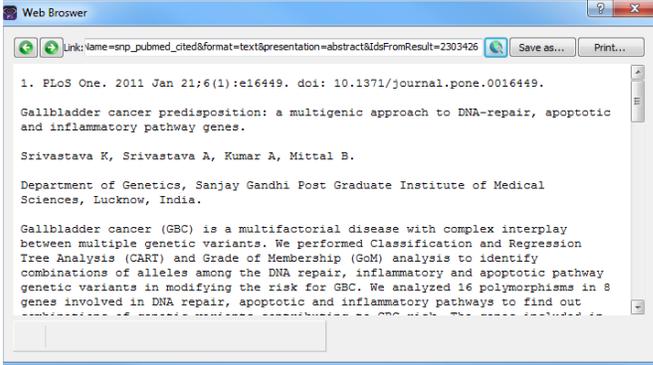
Option	Description
<p>Update Preferred Transcript</p>	<p>Opens the Update Preferred Transcript dialog box. You can select a different preferred transcript for the variant on the Change Transcript To dropdown list.</p> <p><i>Figure 5-32: Update Preferred Transcript dialog box</i></p>  <p>Note: When you select a different transcript here, 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 90. 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.</p>
<p>External - Contains all the sub-menu options for viewing the variant in third-party tools.</p>	
<p>View in Alamut</p>	<p>Opens the variant for viewing in Alamut.</p> <p>Note: To view a variant in Alamut, your Alamut settings must be specified. See “To specify Alamut settings” on page 61.</p>
<p>View in UCSC</p>	<p>Opens the variant for viewing in the UCSC Variant Genome Browser.</p>

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

Option	Description
View PubMed Abstract	<p>Searches PubMed for the variant and displays the appropriate abstract in the PubMed window.</p> <p><i>Figure 5-33: PubMed window</i></p>  <p>You can do the following for the abstract:</p> <ul style="list-style-type: none"> • 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.
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 267.
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 267.
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.
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>

Chapter 5
 Managing Patients and Runs

Table 5-8: 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 modify the run date/time and/or assign the run to a sample group. You specify these settings on a per run basis, or you specify these settings on a per sample basis. At a minimum, you must always load a VCF file. Optionally, you can also load a BAM file to include coverage information for the panel regions.

To add a run

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

The New Run dialog box opens.

Figure 5-34: New Run dialog box

The screenshot shows the 'New Run' dialog box with the following settings:

- Run Name: (empty text field)
- Required Settings:
 - Chemistry: default (dropdown with 'New...' button)
 - Instrument: default (dropdown with 'New...' button)
 - Reference: Human 37 (dropdown)
 - Panel: CANcer panel -Compr. (dropdown with 'New...' button)
 - Panel Group: default (text field)
 - Multiple Samples in VCF: (checkbox)
 - Select Variant and/or Coverage Files... (button)
- Optional Settings:
 - Run Date/Time: 4/28/2016 11:34:18 AM (date/time picker)
 - User Group: Supervisor (dropdown)
 - Sample Group: (dropdown with 'New...' button)
 - Subpanel: (dropdown)
 - Add New Patient... (button)
 - Identify Patient ID from Sample by Separator: (checkbox)
 - Separator: (underscore) (dropdown)
 - Import Pathogenicity from VCF: (checkbox)
 - Submit Hotspots: (checkbox)
 - Accepted VCF filters: All (text field)
 - Edit (button)
- Buttons: OK, Cancel

2. Enter the name for the new run.



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

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Managing Patients and Runs

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 Files	If the correct reference files are not available, then you might need to specify the reference directory and/or add the necessary reference files to the directory. See “Specifying your Geneticist Assistant Settings” on page 53. Note: SoftGenetics provides the necessary references files for your Geneticist Assistant installation. Contact SoftGenetics about downloading these files. Note: This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
Panel	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 85. Note: This is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
Multiple Samples in VCF	Select this option if you are loading a VCF file that contains variants from multiple samples.
Optional Settings	
Run Date/Time	The default value for the Run Date and Time is the current day’s date and time, but you can change one or both values as needed. To change the value, select the value that is to be changed, for example, the month, and then you can manually enter the new value, or you can use the Up/Down field arrows to change the value. Note: You must select each value that is to be changed one at a time. If you set the Run Date/Time here, then this is a run-specific option. You also have the option of setting this option on a per sample basis later in the process.
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 99 .
Identify Patient ID from Sample by Separator	Selected by default. If the file names for the loaded sample files contain the Patient ID, then define how the Patient ID is separated from the rest of the file name, for example, by an underscore (_), a dot (.), and so on. This allows each sample to be linked automatically to the corresponding patient. If the file names for the loaded sample files do not contain the Patient ID, then clear this option.
Import Pathogenicity from VCF	Select this option if you are loading a VCF file that contains pathogenicity information and you want to import this information into the Geneticist Assistant database. 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.
Submit Hotspots	Select to submit any variants that are included in the loaded VCF files that have an allele frequency value = 0 to the Geneticist Assistant database.
Accepted VCF filters	Applicable after you load sample files. See Step 5 .

4. To load your files, do the following:

- Click Select Variant and/or Coverage Files to open the Select Files dialog box.
- In the Select Files dialog box, browse to and select the 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 modify 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.

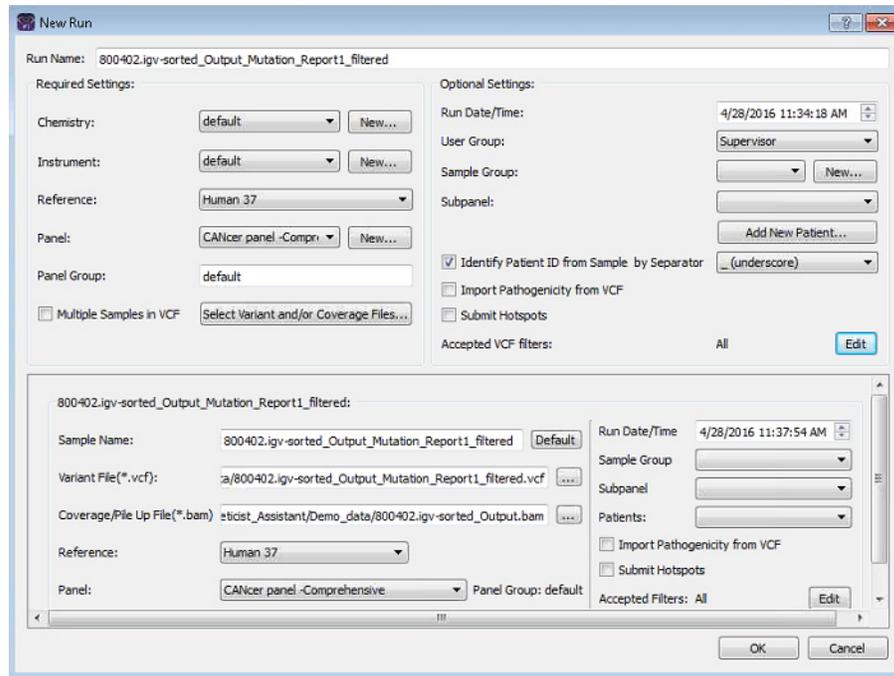
- If you have a *single* sample to load, and the VCF and BAM file names do *not* contain corresponding fields in the file names, then select the VCF or BAM file, and then after you click Open, the file is placed in the appropriate field for the sample entry. You can then click the Browse button  next 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 288.
- By default, the sample name is set to the VCF file name. The name is displayed at the top of the sample entry. You can modify the sample name.
- If you load a sample file in error, then you can right-click the sample name, and then click Remove this sample.

After the files are loaded for a sample, the full directory paths (directory and file name) for the loaded files are displayed in the appropriate fields for each sample entry at the bottom of the New Run dialog box. See [Figure 5-35 on page 153](#).



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.

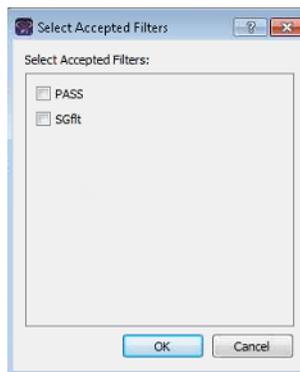
Figure 5-35: New Run dialog box with loaded sample files



- After you load your sample files, you can leave Accepted VCF filters set to the default value of All, or optionally, you can click Edit to open the Select Accepted Filters dialog box, and specify the filters that are available in the loaded VCF file. Only those variants with the selected filter values are imported into Geneticist Assistant.

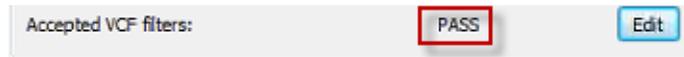
After you select the filters, and then click OK, the Select Accepted Filters dialog box closes. The selected filters replace the default value of All. See [Figure 5-37 on page 154](#).

Figure 5-36: 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.

Figure 5-37: 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 as needed:
 - Modify the sample name.



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

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



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

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



If the correct patient is not available, then you can click Add New Patient to add the patient. See [“Adding Patients” on page 107](#).

- 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, modify 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 155](#).

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 157.](#)
- [“The Runs Tab Context Menu” on page 159.](#)

Chapter 6
Geneticist Assistant Data Review - Runs Tab

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.

Figure 6-1: Runs tab

ID	Name	Run Date Time	Add Date Time	Chemistry	Instrument	# Regions	# Passed Regions	Status
39	Test	6/9/2015 3:35:52 PM	6/9/2015 3:36:08 PM	default	default	154	151	New
36	RainDance Cancer Panel Test 2	4/29/2015 2:16:24 PM	4/29/2015 2:16:58 PM	default	default	194	194	New
32	Demo2	10/19/2014 2:08:22 PM	10/19/2014 2:17:46 PM	default	default	616	608	QC Passed
31	Demo	10/19/2014 1:54:08 PM	10/19/2014 2:04:08 PM	default	default	770	759	Complete
13	Control0625	6/25/2014 1:59:54 PM	7/22/2014 10:08:18 AM	default	default	462	456	New
12	Control0702	7/2/2014 1:46:03 PM	7/22/2014 9:56:05 AM	default	default	462	456	New

You can do the following on the Runs tab:

- Change the display of the Runs tab. See [“To change the Runs tab display”](#) below.
- Modify certain values that are displayed for a run. See [“To modify values for a selected run”](#) on page 158.
- Open a context menu with a variety of options for working with and displaying run-specific data. See [“The Runs Tab Context Menu”](#) on page 159.

To change the Runs tab display

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.

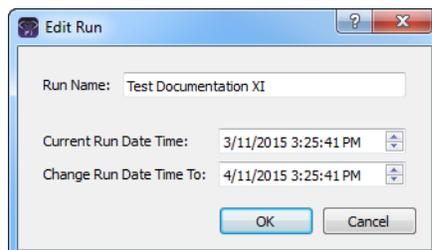
To modify values for a selected run

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 modify it as necessary, and then click out of the field.
- Select a different Chemistry and/or Instrument for the run.

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

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” on page 49.

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 Details	<p>Opens the Run tab. See Chapter 7, “Geneticist Assistant Data Review - Run Tab,” on page 179.</p> <p>Note: You can also double-click a run entry to open the Run tab.</p>
Add All to Custom Report	<p>Selects all the data that is displayed for the run for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267.</p>
Compare Samples	<p>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 “To compare all the samples within a run from the Runs tab” on page 161.</p>
Panel Statistics	<p>Opens the Panel Statistics tab. The tab has options for displaying specific information based on the panel that was used for the run. See “To calculate panel statistics (Runs tab)” on page 163.</p>
Permissions	<p>Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the run. If you are an Administrator user, then you can set or modify the permissions for a run that any user has submitted. If you are a standard user, then you can set or modify the permissions for only those runs that you have submitted to the Geneticist Assistant database, or for those runs that other users have submitted and to which you have been granted Can Read and Can Write permissions. See “To set or modify the Read/Write permissions for a run” on page 168.</p>
Edit Run	
Add Samples to Run	<p>Opens the New Run dialog box so that you can add more samples to the selected run. See “Adding a Run” on page 149.</p>

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

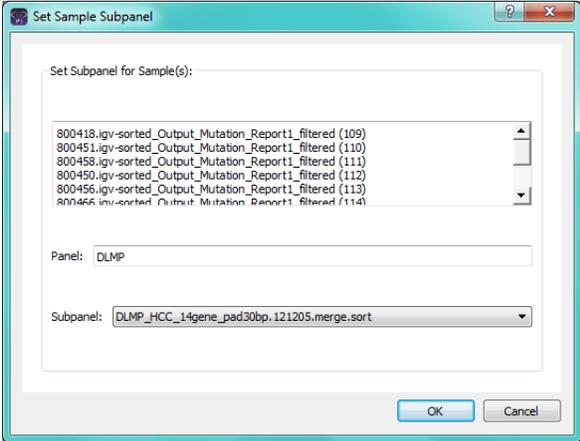
Option	Description
<p>Set Subpanels of Selected Runs</p>	<p>Applicable only if you select a run for which a subpanel was not selected during the setup of the run. CTRL-click to select multiple runs. If you select multiple runs, then all the runs must use the same panel. Opens the Select a Subpanel dialog box. The dialog box displays the samples and the panel for the selected runs and a Subpanel dropdown list with all the available subpanels.</p> <p><i>Figure 6-3: Set Sample Subpanel dialog box</i></p>  <p>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.</p>
<p>Delete Run</p>	<p>Delete the selected run from the Geneticist Assistant database.</p>
<p>Exports</p>	
<p>Export to VCF</p>	<p>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.</p>
<p>Save Sample Reports</p>	<p>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. You must specify a location in which to save the reports. Each report is named the same as the sample and the report format is HTML, and you cannot change either of these values.</p> <p>Note: For more information about standard reports, see Chapter 10, “Geneticist Assistant Reports,” on page 267.</p>
<p>Create Batch Variants Report</p>	<p>Opens the Batch Variants Report dialog box for generating the Batch Variants report for all the samples in a selected run. See “To manage a Batch Variants report (Runs tab)” on page 170.</p>
<p>Create Batch Coverage Report</p>	<p>Opens the Batch Coverage Report dialog box for generating the Batch Coverage report for all the samples in the selected run or runs. See “To generate a Batch Coverage report (Runs tab)” on page 175.</p>

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

Option	Description
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>
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 .

To compare all the samples within a run from the Runs tab

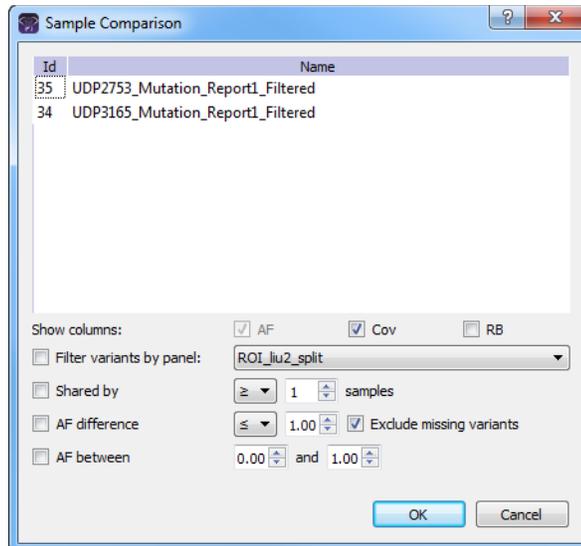


To compare only selected samples within a run, see [“To compare selected sample data for a run from the Run tab” on page 188](#).

1. 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 run are displayed in the dialog box. See [Figure 6-4 on page 162](#).

Figure 6-4: 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. Coverage (Cov) is displayed by default, but you can clear this selection. Read Balance (RB) is not displayed by default, but you can select this option.
Filter variants by panel	Limits the variants that are displayed on the Sample Comparison tab to only those variants that are contained within the selected panel.
Shared by	Limits the variants that are displayed on the Sample Comparison tab 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 in a number of samples that does not meet this criteria, then the variant is not displayed in the results.
AF difference	Limits the variants that are displayed on the Sample Comparison tab to only those variants that have a difference in allele frequency as specified by the operator that is selected on the dropdown list (>, <, or =) and the number of samples. If a variant is found with an allele frequency difference that does not meet this criteria, then the variant is not displayed in the results.
<ul style="list-style-type: none"> Exclude missing variants 	Selected by default. Regardless of the allele frequency difference, always display variants that are <i>not</i> 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 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. A distinct gray vertical line separates the two sides of the tab. The left 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 right side of the tab shows the variant details.

Figure 6-5: Sample Comparison tab

To calculate panel statistics (Runs tab)

The Panel Statistics tab has options for **displaying** specific information based on a selected panel and panel group as well as any or all of the following criteria:

- All 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.
- Selected panel samples.

To display data on the Panel Statistics tab

1. Select the appropriate run, or CTRL-click to select multiple runs on the Runs tab.
2. Right-click any selected run, and on the context menu that opens, click Panel Statistics.



If you select 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.

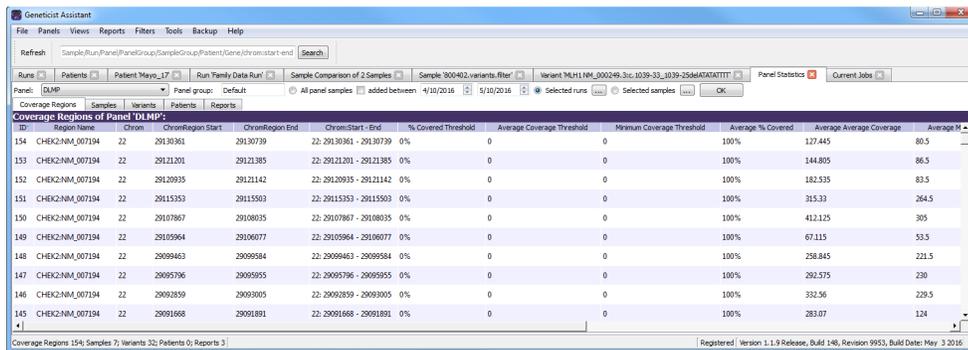
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Geneticist Assistant Data Review - Runs Tab

The Panel Statistics tab opens. The Coverage Regions sub-tab is the open tab. The panel and panel group that were used for the run are selected. 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. By default, the statistics are calculated for all the samples in the selected run that have been added to the panel.



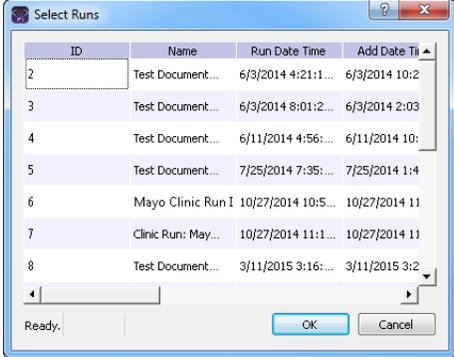
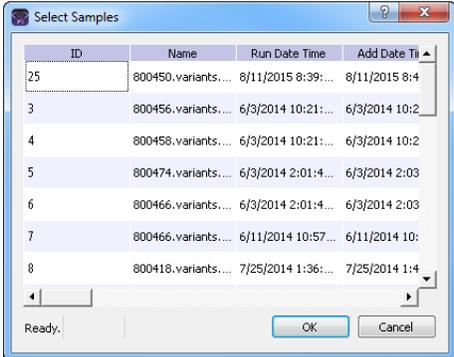
If the run contains a small number of samples, such as two to four, then, by default, the panel statistics are calculated based only on the samples for the run. Before the statistics are calculated, a Very few samples message dialog box opens, warning you of this and asking you if you want to continue. You can click Yes, or you can click Cancel to return to the Runs tab.

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



- Optionally, modify 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	<p>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:</p> <ul style="list-style-type: none"> 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
<p>Selected runs</p>	<p>Selected by default. To select specific runs for the panel, choose Selected runs, and then click the Browse button  that is next to Selected runs to open the Select Runs dialog box. All the runs for the selected panel are displayed in the dialog box. Select the appropriate run, or CTRL-click to select multiple runs, and then click OK.</p> <p><i>Figure 6-7: Select Runs dialog box</i></p> 
<p>Selected samples</p>	<p>To select only specific samples for the panel, choose Selected samples and then click the Browse button  that is next to Selected samples to open the Select Samples dialog box. All the samples for the selected panel are displayed in the dialog box. Select the appropriate sample, or CTRL-click to select multiple samples, and then click OK.</p> <p><i>Figure 6-8: Select Samples dialog box</i></p> 

4. 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 166.
 - Samples sub-tab. See [“Samples sub-tab \(Panel Statistics tab\)”](#) on page 166.
 - Variants sub-tab. See [“Variants sub-tab \(Panel Statistics tab\)”](#) on page 167.

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- Patients sub-tab. See “Patients sub-tab (Panel Statistics tab)” on page 167.
- Reports sub-tab. See “Reports sub-tab (Panel Statistics tab)” on page 168.

Coverage Regions sub-tab (Panel Statistics tab)

The Coverage Regions sub-tab has a single pane, the Coverage Regions pane, that lists by default, the coverage information across all the samples for the selected run or runs for every region in the selected panel and panel group. See “Coverage Regions tab” on page 192 for a detailed discussion of the information that is displayed on this sub-tab.

Figure 6-9: Coverage Regions sub-tab (Panel Statistics tab)

ID	Region Name	Chrom	Chrom:Region Start	Chrom:Region End	Chrom:Start - End	% Covered Threshold	Average Coverage Threshold	Minimum Coverage Threshold	Average % Covered	Average Coverage	Average M
154	CHEK2-NM_007194	22	29130361	29130739	22: 29130361 - 29130739	0%	0	0	100%	127.445	80.5
153	CHEK2-NM_007194	22	29121201	29121385	22: 29121201 - 29121385	0%	0	0	100%	144.805	86.5
152	CHEK2-NM_007194	22	29120935	29121142	22: 29120935 - 29121142	0%	0	0	100%	182.535	83.5
151	CHEK2-NM_007194	22	29115353	29115503	22: 29115353 - 29115503	0%	0	0	100%	315.33	264.5
150	CHEK2-NM_007194	22	29107867	29108035	22: 29107867 - 29108035	0%	0	0	100%	412.125	305
149	CHEK2-NM_007194	22	29105964	29106077	22: 29105964 - 29106077	0%	0	0	100%	67.115	53.5
148	CHEK2-NM_007194	22	29099463	29099584	22: 29099463 - 29099584	0%	0	0	100%	258.845	221.5
147	CHEK2-NM_007194	22	29095796	29095955	22: 29095796 - 29095955	0%	0	0	100%	292.575	230
146	CHEK2-NM_007194	22	29092859	29093005	22: 29092859 - 29093005	0%	0	0	100%	332.56	229.5
145	CHEK2-NM_007194	22	29091668	29091891	22: 29091668 - 29091891	0%	0	0	100%	283.07	124

Samples sub-tab (Panel Statistics tab)

The Samples sub-tab has a single pane, the Samples pane, that lists by default, all the samples that were imported in the selected run or runs 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 215.) 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 184.

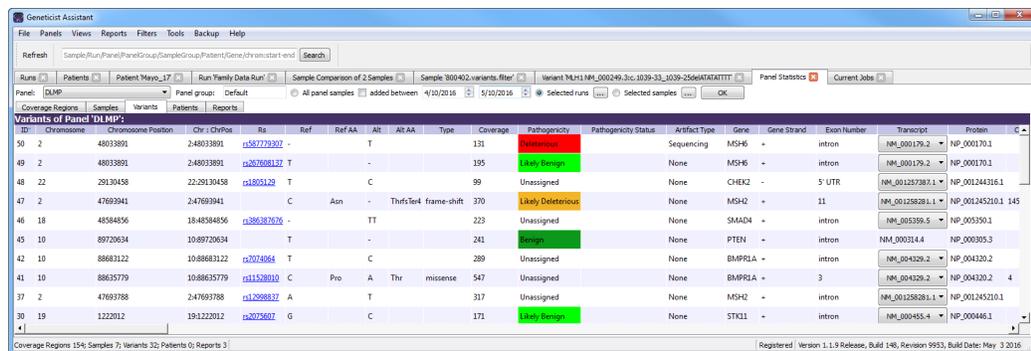
Figure 6-10: Panel Statistics tab, Samples sub-tab

ID	Name	Run Date Time	Add Date Time	Run	Panel	Panel Group	Reference	Sample Groups	# Regions	# Regions Passed	Gc Content	Patient External ID	Status	Missed Clinical Variants	C:\Users\Spec
21	800402.variants.filter	4/7/2015 10:07:28 A...	4/7/2015 10:07:49 A...	Test Document...	DLMP	Default	Human_37_s92	154	0	0	N/A	N/A	N/A	C:\Users\Spec	
20	800418.variants.filter	4/7/2015 9:42:44 AM	4/7/2015 9:49:20 AM	Test Document...	DLMP	Default	Human_37_s92	154	0	0	N/A	N/A	N/A	C:\Users\Spec	
19	Sample_s3_738201_Ou...	4/7/2015 8:41:26 AM	4/7/2015 8:41:56 AM	Test Document...	DLMP	Default	Human_37_s92	154	0	0	N/A	N/A	N/A	C:\Users\Spec	
18	Sample_s3_738201_Ou...	4/7/2015 8:41:26 AM	4/7/2015 8:41:55 AM	Test Document...	DLMP	Default	Human_37_s92	154	0	0	N/A	N/A	N/A	C:\Users\Spec	
17	Sample_s2_749888_Ou...	4/7/2015 8:41:26 AM	4/7/2015 8:41:54 AM	Test Document...	DLMP	Default	Human_37_s92	154	0	0	N/A	N/A	N/A	C:\Users\Spec	
16	800418.variants.filter	3/25/2015 10:59:07 ...	3/25/2015 11:05:35 ...	800402.variants...	DLMP	Default	Human_37_s92 Mayo Test 1	143	0	0	0	Review...	N/A	C:\Users\Spec	
15	800402.variants.filter	3/25/2015 10:59:07 ...	3/25/2015 11:05:31 ...	800402.variants...	DLMP	Default	Human_37_s92 Mayo Test 1	143	0	0	0	New	N/A	C:\Users\Spec	

Variants sub-tab (Panel Statistics tab)

The Variants sub-tab has a single pane, the Variants pane, that lists by default, all the variants that were detected in all the samples that were imported in the selected run or runs 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 245.) 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 218.)

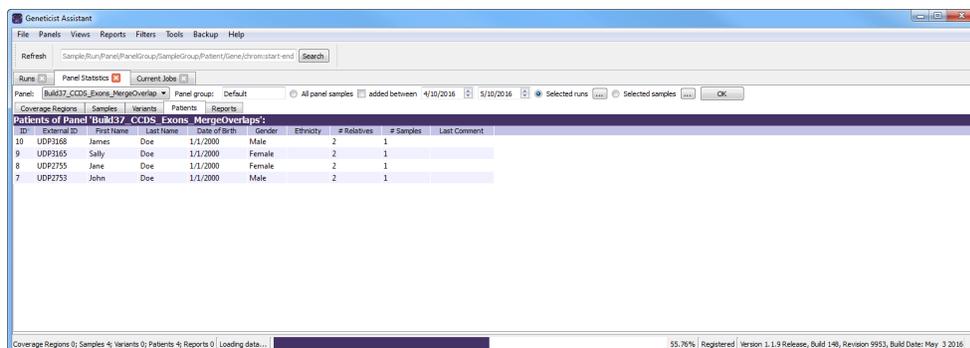
Figure 6-11: Panel Statistics tab, Variants sub-tab



Patients sub-tab (Panel Statistics tab)

The Patients sub-tab has a single pane, the Patients pane, that lists by default, all the patients who were associated with all the samples 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 112.

Figure 6-12: Panel Statistics tab, Patients sub-tab



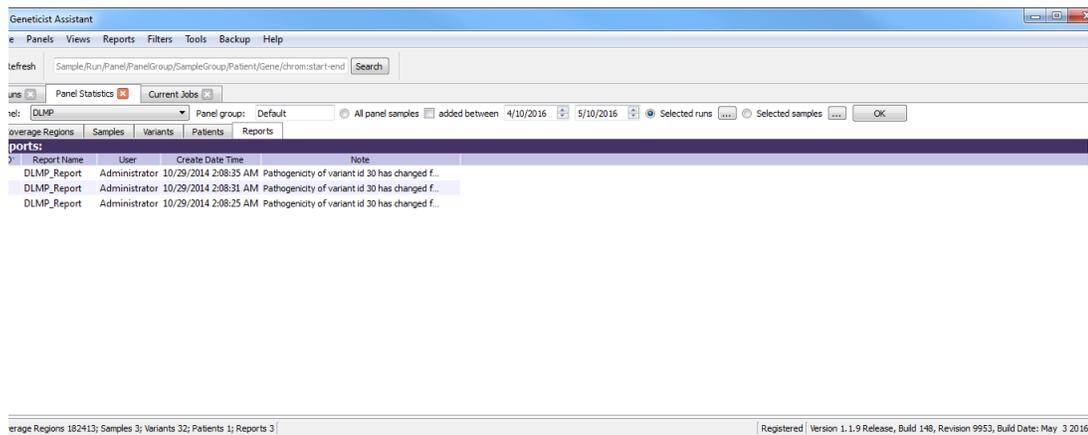
Reports sub-tab (Panel Statistics tab)

The Reports sub-tab contains a single pane, the Reports pane, that lists by default, all the reports that were generated for the selected run or runs 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 6-13: 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 126](#).



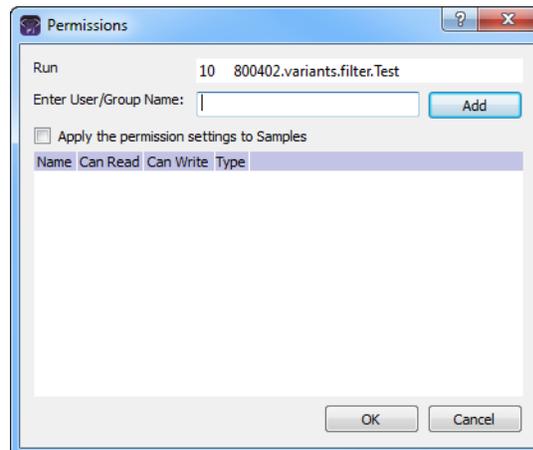
For detailed information about generating and saving Panel Statistics reports, see [Chapter 10, “Geneticist Assistant Reports,” on page 267](#).

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

1. If you have not already done so, open the Runs tab.
2. Select the run, or CTRL-click to select the multiple runs for which the permissions are being set.
3. Right-click any selected run, and on the context menu that opens, click Permissions.
The Permissions dialog box opens. See [Figure 6-14 on page 169](#).

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

Figure 6-14: Permissions dialog box (Runs tab)



4. Modify the Read and Write run permissions for any user or group as needed and then go to [Step 8](#); otherwise, to add run permissions for a new user or group, go to [Step 5](#).



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.

5. 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.



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.

6. Select the appropriate 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 Read permission is already assigned for the selected user or group. If just this permission is selected, then the user or group can simply view the run. They cannot modify it.
7. Optionally, if the user or group must be able to modify the run as well, then select the Can Write permission.
8. 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.



You can assign Read and Write permissions for the samples in a run on a per sample basis. See [“To set or modify the Read/Write permissions for the samples in a run on a per sample basis”](#) on page 202.

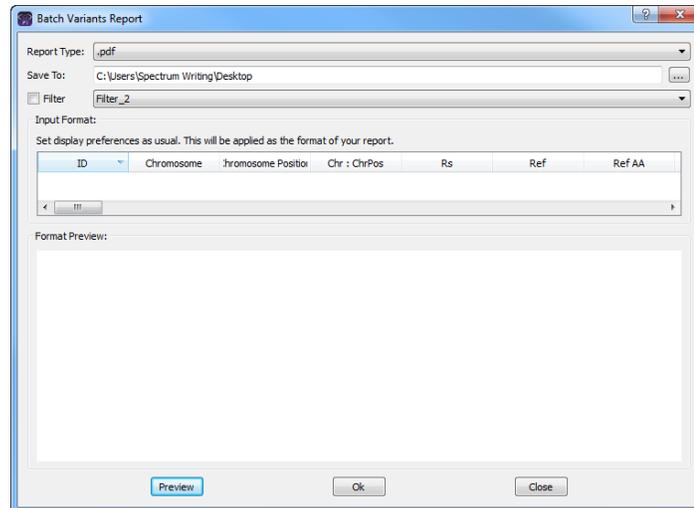
9. Click OK.
A Permission Updated message opens.
10. Click OK.
Both the message and the Permissions dialog box close. The specified permissions are set for the runs.

To manage a Batch Variants report (Runs tab)

A *Batch Variants report* is a report that includes information for variants in a selected sample, and allows you to create multiple reports for multiple samples in a single step. If you generate a Batch Variants report from the Runs tab, then a separate report is saved for each sample of the selected run. A *Batch Variants report configuration* determines the data that is to be included 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 (PDF, HTML and so on), or the location in which the report is to be saved. Instead, you must specify the format and location in which to save the report every time you generate a report for a new or saved configuration. You can generate a Batch Variants report according to a new or saved configuration, modify a Batch Variants report configuration, or delete a Batch Variants report configuration. See:

- [“To generate a Batch Variants report based on a new configuration”](#) on page 171.
- [“To generate a Batch Variants report based on a saved configuration”](#) on page 172.
- [“To modify a Batch Variants report configuration”](#) on page 174.
- [“To delete a Batch Variants Report configuration”](#) on page 175.

Figure 6-15: Batch Variants Report dialog box



To generate the report for all variants in only selected samples for the run, see “To manage a Batch Variants report (Run tab)” on page 204.

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 the first sample in the list of samples 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.

1. If you have not already done so, open the Runs tab.
2. Select the run or CTRL-click to select the multiple runs for which the Batch Variant report is to be generated.
3. Right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, on the Report Type dropdown list, select the report type. (The default value is PDF.)
5. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
 - Leave this location as-is, and then go to [Step 6](#).
 - 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.

6. Optionally, to filter the data that is to be displayed in the report, click 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.

7. Optionally, to set the display preferences for the report, do any of the following as needed:
 - 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 each of the samples in the selected run. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.

10. Click OK.

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

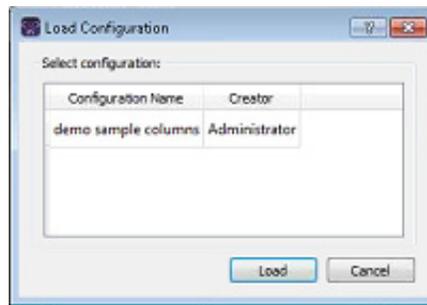
To generate a Batch Variants report based on a saved configuration

1. If you have not already done so, open the Runs tab.
2. Select the run or CTRL-click to select the multiple runs for which the Batch Variant report is to be generated.
3. Right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, select the output format for the report.

5. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
 - Leave the location as-is, and then go to [Step 6](#).
 - 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.
6. Right-click on any heading in the Input Format pane, and then choose Select Columns. The Select Columns dialog box opens.
7. Click Load Configuration.

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

Figure 6-16: Load Configuration dialog box



8. Select a configuration, and then click Load.

The Load Configuration dialog box closes. The Select Columns dialog box remains open. The dialog box now reflects the columns that are to be included in the report based on the selected configuration.
9. Click Close (x) to close the Select Columns dialog box.
10. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.
11. Click OK.

The Batch Variants report is generated for for each of the samples in the selected run. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.
12. Click OK.

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

To modify a Batch Variants report configuration

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

1. If you have not already done so, open the Runs tab.
2. Select the run or CTRL-click to select the multiple runs for which the Batch Variant report is to be generated.
3. Right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants 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, this field is blank.
5. Do one of the following:
 - Leave this location as-is, and then go to [Step 6](#).
 - 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.
6. Right-click on any heading in the Input Format pane, and then choose Select Columns. The Select Columns dialog box opens.
7. To modify the report configuration, see [“To create/modify a pane configuration” on page 39](#).
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 each of the samples in the selected run. After the report is successfully generated, a Batch Variants Report Finished dialog box opens, with a message that indicates where the report was saved.

10. Click OK.

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

To 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. If you have not already done so, open the Runs tab.
2. Select the run or CTRL-click to select the multiple runs for which the Batch Variant report is to be generated.
3. Right-click any selected run, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, right-click on any heading in the Input Format pane, and then choose Select Columns.
The Select Columns dialog box opens.
5. See [“To delete a pane configuration” on page 41](#).
6. Click Close (x) to close the Batch Variants Report dialog box.

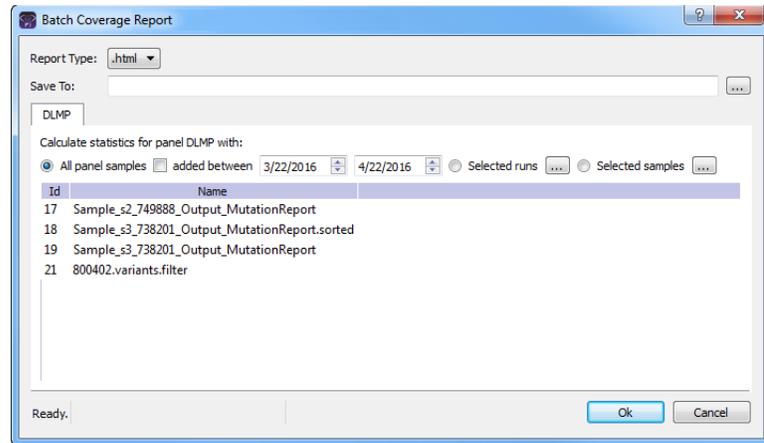
To generate a Batch Coverage report (Runs tab)

The Batch Coverage report saves a coverage report for each of the samples for a selected run or runs in a specific panel or panels and panel group. The report also includes coverage statistics for the panel that are calculated based on one of the following options:

- All 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.
 - Selected panel samples.
1. If you have not already done so, open the Runs tab.
 2. Select a single run, or CTRL-click to select multiple runs.
 3. Right-click on any selected run, and on the context menu that opens, click Export > Create Batch Coverage report.

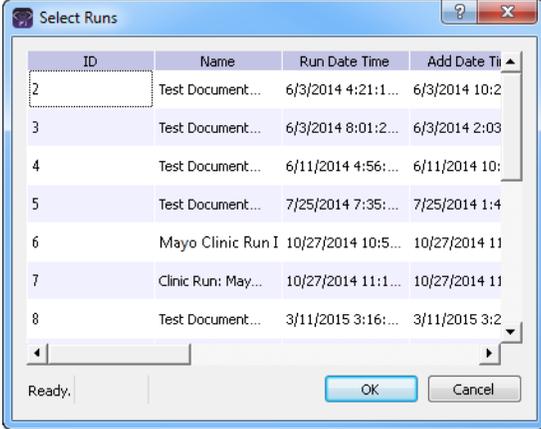
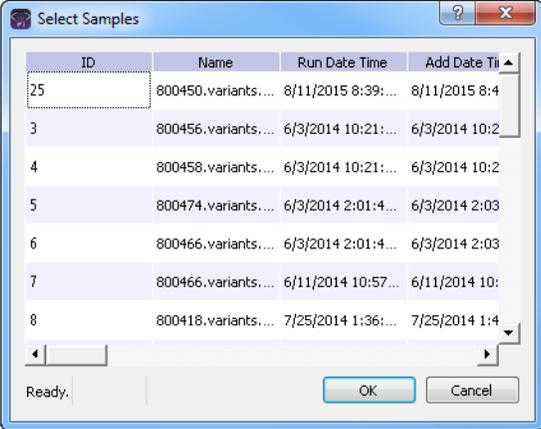
The Batch Coverage Report dialog box opens. All the samples in the selected run are displayed in the bottom pane of the dialog box. If multiple runs are selected with different panels, then a separate tab is displayed for each panel. See [Figure 6-17 on page 176](#).

Figure 6-17: Batch Coverage Report dialog box (Runs tab)



4. Leave the report type set to the default value of HTML, or select a different report type.
5. 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 reports.
6. 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.

Option	Description
All panel samples added between	<p>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:</p> <ul style="list-style-type: none"> • 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
<p>Selected runs</p>	<p>To select specific runs for calculating the panel coverage statistics, choose Selected runs, and then click the Browse button  that is next to Selected runs to open the Select Runs dialog box. All the runs for the panel are displayed in the dialog box. Select the appropriate run, or CTRL-click to select multiple runs, and then click OK.</p> <p><i>Figure 6-18: Select Runs dialog box</i></p> 
<p>Selected samples</p>	<p>To select only specific samples for calculating the panel coverage statistics, choose Selected samples and then click the Browse button  that is next to Selected samples to open the Select Samples dialog box. All the samples for the panel are displayed in the dialog box. Select the appropriate sample, or CTRL-click to select multiple samples, and then click OK.</p> <p><i>Figure 6-19: Select Samples dialog box</i></p> 



If the panel coverage statistics are to be calculated 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 Runs tab.

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 for each sample in the selected run or runs in the specified format and the specified location.

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 181.](#)
- [“Samples pane \(Run tab\)” on page 182.](#)
- [“Coverage Regions tab” on page 192.](#)
- [“The Samples Pane \(Run tab\) Context Menu” on page 184.](#)
- [“Run’s Status Changes pane” on page 212.](#)
- [“Reports pane \(Run tab\)” on page 213.](#)

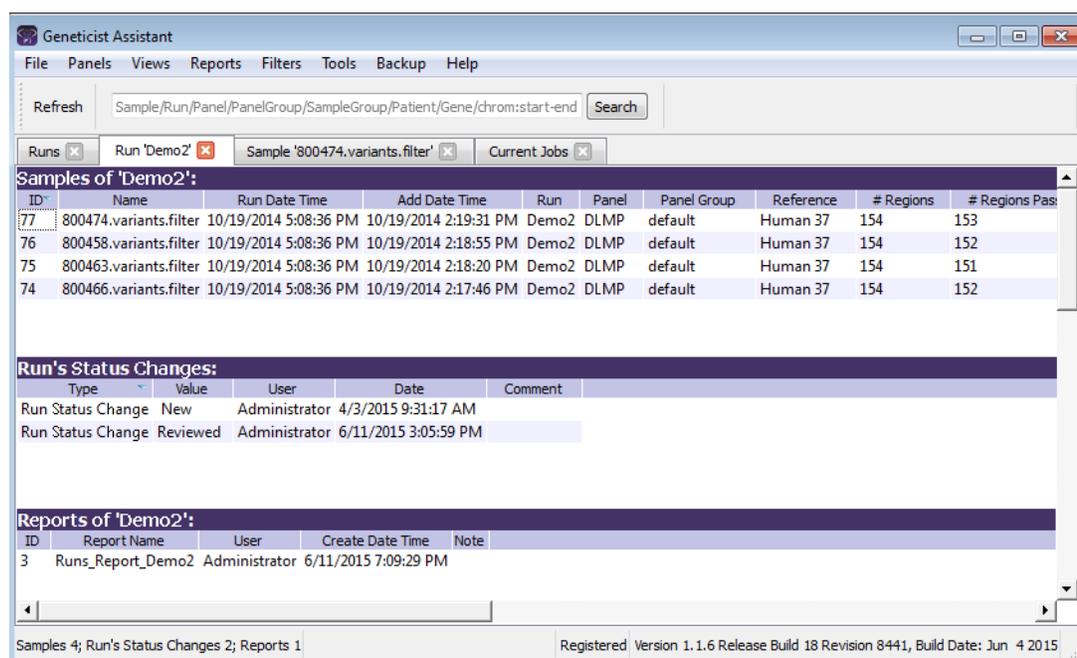
Chapter 7
Geneticist Assistant Data Review - Run Tab

Overview of the Run Tab

The Run tab displays information for a run that was selected on the Runs tab. To open the Run tab, do one of the following:

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

Figure 7-1: Run tab



The Run tab has three panes. From top to bottom, these three panes are the following:

- The Samples pane. See [“Samples pane \(Run tab\)”](#) below.
- The Run’s Status Changes pane. See [“Run’s Status Changes pane”](#) on page 212.
- The Reports pane. See [“Reports pane \(Run tab\)”](#) on page 213.

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 182](#).
- Determining missed clinical variants. See [“To determine missed clinical variants for a sample” on page 183](#).
- Using the context menu. See [“The Samples Pane \(Run tab\) Context Menu” on page 184](#).

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.

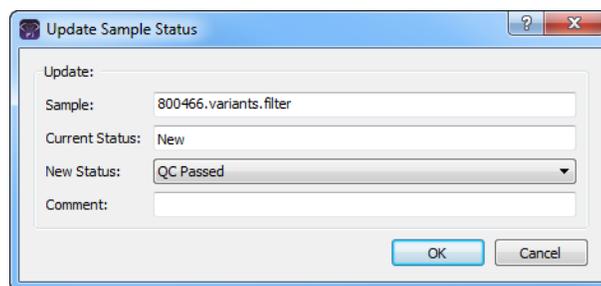
1. If you have not done so already, open the Run tab. (See [“Overview of the Run Tab” on page 181](#).)
2. On the Samples pane, on the Status dropdown list, select a different status for the sample.



You can select a different value from one of the available default values (New, QC Passed, Reviewed, or Complete), or if needed, you can create custom statuses, and then select one of these custom statuses instead. See [“Managing Objects” on page 49](#).

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” on page 238](#).

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.

To determine missed clinical variants for a sample

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

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

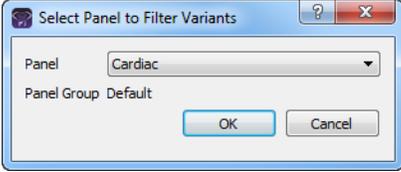
Option	Description
Compare Samples	Displayed only after two or more samples are selected. Opens the Sample Comparison dialog box. You use the options on this dialog box to compare the coverage, frequency, and read balance information for each sample that is being compared. See “To compare selected sample data for a run from the Run tab” on page 188.
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 190.
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 215.
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 192.
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 “To calculate panel statistics (Run tab)” on page 197.
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 267.
Show Sample Variants Filtered by Panel	<p>Displays all the variants in the selected sample filtered by a selected panel. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected run.</p> <p><i>Figure 7-3: Select Panel to Filter Variants dialog box</i></p> 
Permissions	Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for the sample. If you are an Administrator user, then you can set or modify the permissions for any sample in any run that any user has submitted. If you are a standard user, then you can set or modify the permissions for only those samples that are contained in runs that you have submitted to the Geneticist Assistant database, or for those samples that are contained in runs that were submitted by other users and to which you have been granted Can Read and Can Write permissions. See “To set or modify the Read/Write permissions for the samples in a run on a per sample basis” on page 202.

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

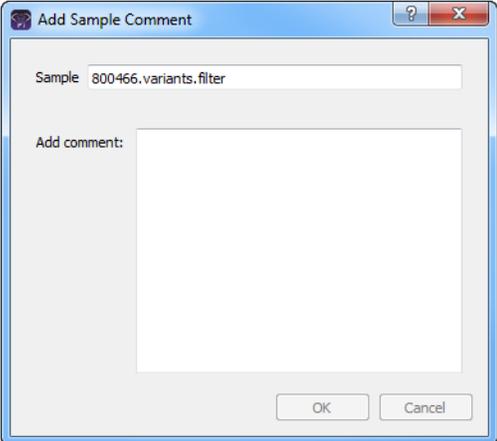
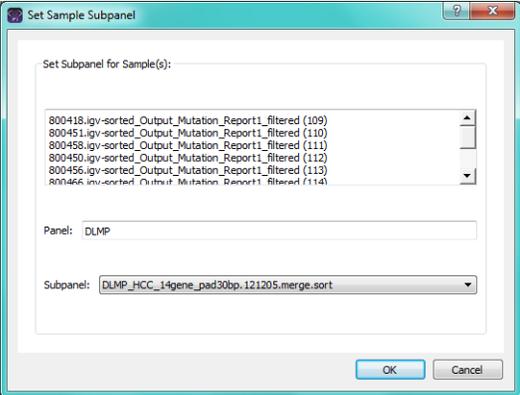
Option	Description
<p>Edit Sample</p> <p>Add Comment</p>	<p>Opens the Add Sample Comment dialog box. You add a comment for the associated sample in the Add Comment field. The comment is displayed in the Comments field on the Samples sub-tab on the Patient tab, the Comments field in the Samples pane on the Run tab, and in the Comments pane on the Sample Data sub-tab on the Sample tab.</p> <p><i>Figure 7-4: Add Sample Comment dialog box</i></p>  <p>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.</p>
<p>Set Subpanel</p>	<p>You must select a sample or samples for which a subpanel was not selected during the setup of the run. 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.</p> <p><i>Figure 7-5: Set Sample Subpanel dialog box</i></p>  <p>Select a subpanel on the Subpanel dropdown list, and then click OK. The subpanel is applied to sample or samples.</p>

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

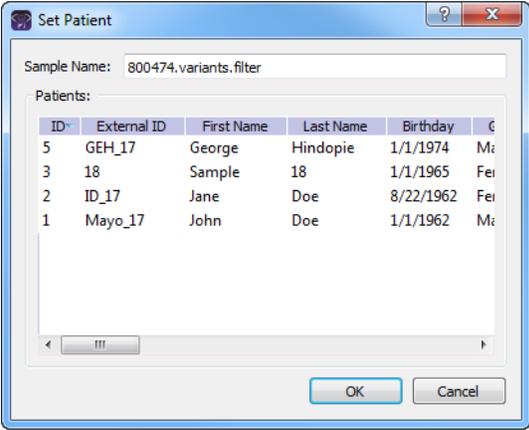
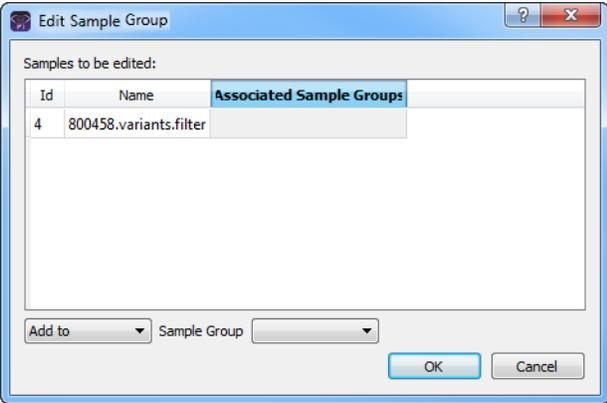
Option	Description
Set Patient	<p>Opens the Set Patient dialog box. Select the patient that is to be associated with the sample, and then click OK.</p> <p><i>Figure 7-6: Set Patient dialog box</i></p>  <p>Note: You also have the option of associating a patient with a sample from the Sample tab. See “To associate a patient with a sample” on page 242.</p>
Edit Sample Groups	<p>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.</p> <p>Note: If the appropriate sample group is not available, you can add it. See “Managing Objects” on page 49.</p> <p><i>Figure 7-7: Edit Sample Group dialog box</i></p> 
Delete Sample	Deletes the sample from the run.
Exports	
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.

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

Option	Description
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 (HTML) 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 HTML, and you cannot change either of these values. Note: For more information about standard reports, see Chapter 10, “Geneticist Assistant Reports,” on page 267.
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 “To manage a Batch Variants report (Run tab)” on page 204.
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 “To generate a Batch Coverage report (Run tab)” on page 208.
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 37. 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 58.
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.

To compare selected sample data for a run from the Run tab

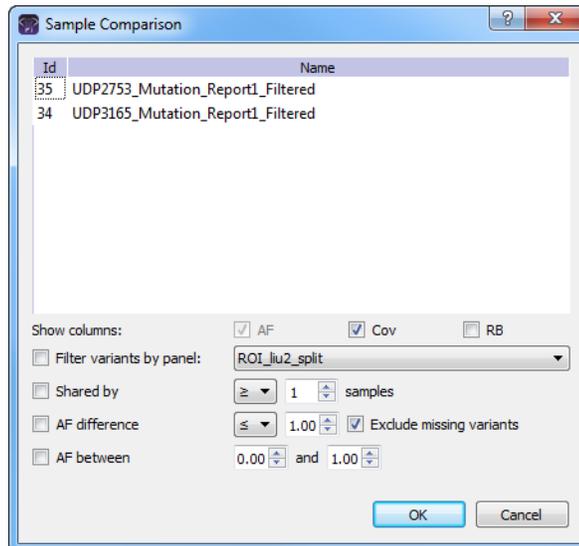


To compare all samples within a run, see *“To compare all the samples within a run from the Runs tab”* on page 161.

1. If you have not already done so, open the Run tab. (See *“Overview of the Run Tab”* on page 181.)
2. For the appropriate run, CTRL-click to select the samples that are to be compared, and then right-click on 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 7-8: Sample Comparison dialog box



3. 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. Coverage (Cov) is displayed by default, but you can clear this selection. Read Balance (RB) is not displayed by default, but you can select this option.
Filter variants by panel	Limits the variants that are displayed on the Sample Comparison tab to only those variants that are contained within the selected panel.

Option	Description
Shared by	Limits the variants that are displayed on the Sample Comparison tab 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 in a number of samples that does not meet this criteria, then the variant is not displayed in the results.
AF difference	Limits the variants that are displayed on the Sample Comparison tab to only those variants that have a difference in allele frequency as specified by the operator that is selected on the dropdown list (>, <, or =) and the number of samples. If a variant is found with an allele frequency difference that does not meet this criteria, then the variant is not displayed in the results.
<ul style="list-style-type: none"> Exclude missing variants 	Selected by default. Regardless of the allele frequency difference, always display variants that are <i>not</i> 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 that falls within the indicated range in at least one of the compared samples.

4. Click OK.

The Sample Comparison dialog box closes. The Sample Comparison results are displayed on the Sample Comparison tab. A distinct gray vertical line separates the two sides of the tab. The left side of the tab shows the sample-specific information, identifying each sample as 1, 2, and so on. If you hover your cursor over the column heading, then the full sample name is displayed. The right side of the tab shows the variant details.

Figure 7-9: Sample Comparison tab

1:AF	1:Cov	2:AF	2:Cov	3:AF	3:Cov	ID	Chromosome	Chromosome Position	Chr / ChrPos	Ref	Ref AA	Ref AA	Alt	Alt AA
0.322	195	0.322	195			49	2	48033891	2:48033891	r36772311	T	-	-	
0.553	99	0.553	99			48	22	29139458	22:29139458	r1805129	T	-	C	
0.821	370	0.821	370			47	2	47693941	2:47693941	C	Ain	-	Thr/Ter	
0.256	223	0.256	223			46	18	48584856	18:48584856	r38638767	-	-	TT	
0.239	241	0.239	241			45	10	89720634	10:89720634	T	-	-	-	
0.504	289	0.504	289			42	10	88683122	10:88683122	r3712664	T	-	C	
1.119	547	1.119	547			41	10	88635779	10:88635779	r11528010	C	Pro	A	Thr
0.645	317	0.645	317			37	2	47697788	2:47697788	r12299837	A	-	T	
0.491	171	0.491	171	0.477	186	30	19	1222012	19:1222012	r3727607	G	-	C	
				0.628	308	29	14	75515668	14:75515668	r12675881	T	Lys	G	Gln
				0.275	226	28	10	89720634	10:89720634	r34002472	-	-	T	-
				1.180	345	27	5	112155046	5:112155046	G	-	-	A	-
				1.012	504	26	3	37002174	3:37002174	r183922366	TTC	-	-	-
				0.122	37	25	3	37067100	3:37067100	TTT	-	-	-	-
				0.796	375	24	3	30713126	3:30713126	r11496312	T	-	A	-
				0.791	387	22	2	47703500	2:47703500	r2203428	T	-	C	-
0.737	302	0.737	302	0.716	347	21	2	47693959	2:47693959	r3732483	G	-	A	-
				0.731	364	20	2	47666871	2:47666871	r12224300	T	-	C	-
2.101	532	2.101	532			18	17	6354491	17:6354491	r1246308	G	Pro	A	Ser
1.078	264	1.078	264	1.075	258	17	17	63533789	17:63533789	r2915236	T	Pro	C	Pro

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.
2. In the Samples pane, CTRL-click to select the family data that is to be compared.

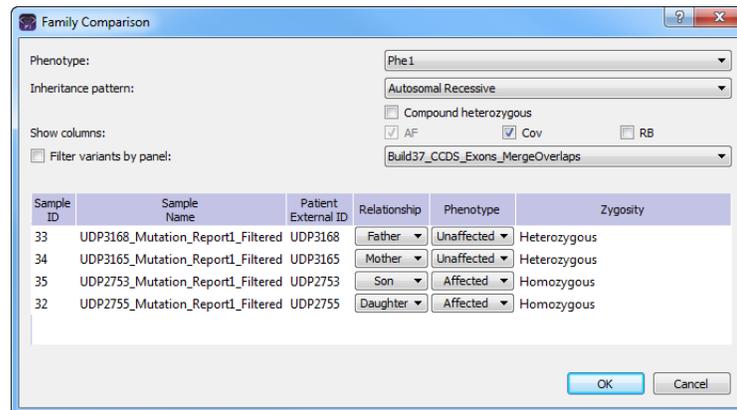


At a minimum, you must select two parent samples and one child sample. You can always select more than one child sample.

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.

Figure 7-10: Family Comparison dialog box



4. Do any or all of the following as needed:
 - 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 a Mother is Unaffected and a Father is 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.

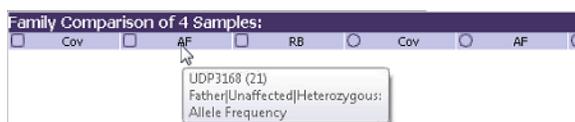
5. Click OK.

The family data comparison is carried out. The Family Comparison tab opens. A distinct gray vertical line divides the two sides. 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](#) below and [Figure 7-12 on page 192](#).



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



Chapter 7
Geneticist Assistant Data Review - Run Tab

Figure 7-12: Family Comparison Results pane

ID	Chr	Chrom	Start	End	% Covered	Average Coverage	Minimum Coverage	Status	Gene															
9	1	1	8425	1400	9	0.556	1.000	8	0.759	1.000	5	1.000	0.620	291	15249789	rs12124212	PRAMEF34	NM_002454813.2:12927-C	synonymous	Passed	Pending	None	+	3
8	1	1	8425	1400	9	0.556	1.000	8	0.759	1.000	5	1.000	0.620	292	15249790	rs12124211	PRAMEF34	NM_002454813.2:13073-A	missense	Miss Chromosome	Pending	None	+	3
9	1	1	8527	1400	58	0.440	1.000	56	1.000	1.000	44	0.984	1.000	955	158097727	rs2528131	WUJ	NM_006113.4:4932A-T	missense	Unassigned	None	None	+	8
166	6	6	494	1400	129	0.431	1.000	121	1.000	1.000	127	0.984	1.000	3036	151444399	rs1164025	APB1	NM_002339512.3:319C-T	synonymous	Passed	Pending	None	-	4
107	4	4	1567	1400	7	0.571	1.000	200	0.879	1.000	73	0.863	1.000	1205	155238222	rs1645614	HRB1	NM_001899312.3:1783A-G	missense	Unassigned	Pending	None	+	3
11	6	6	638	9310	24	0.647	0.718	22	0.773	0.890	10	0.800	1.000	2547	231138406	rs1113155	RPG6	NM_002339313.3:1560A-G	synonymous	Miss Chromosome	Pending	None	+	12
72	6	6	625	1400	79	0.529	1.000	68	1.000	1.000	82	1.000	1.000	3098	235961616	rs12125	DAP11	NM_002197812.3:280A-G	missense	Miss Chromosome	Pending	None	+	3
50	6	6	625	1400	39	0.740	1.000	63	1.000	1.000	53	1.000	1.000	3781	236093195	rs1212663	TRAC1	NM_023394.2:203C-T	synonymous	Unassigned	None	+	14	
81	6	6	586	1400	48	0.396	1.000	52	0.961	1.000	66	1.000	1.000	47133	2389142407	rs2586271	TPPI	NM_006287.4:428+555A-G	missense	Unassigned	None	None	+	intron
133	6	6	474	1400	100	0.540	1.000	104	1.000	1.000	119	0.992	1.000	3099	2302624256	rs12122146	TRAC2	NM_023449.2:424C-A	missense	Unassigned	None	None	+	5
12	4	4	409	1400	19	0.720	0.900	7	1.000	0.470	11	1.000	0.920	3099	2360931208	rs12125211	NOB1B	NM_023794.2:105C-T	missense	Unassigned	None	None	+	5
121	4	4	471	1400	85	0.540	1.000	97	1.000	1.000	79	1.000	1.000	2943	2310557388	rs1212613	MAPP1	NM_002374.3:486C-T	synonymous	Unassigned	None	None	+	7
139	6	6	485	1400	122	0.426	1.000	135	1.000	1.000	118	0.992	1.000	2944	2310557542	rs1212602	MAPP2	NM_002374.3:448C-A	synonymous	Unassigned	None	None	+	7
103	6	6	466	1400	113	0.460	1.000	76	0.907	1.000	74	0.959	1.000	2945	2310557612	rs1212601	MAPP1	NM_002374.3:280A-A	synonymous	Unassigned	None	None	+	7
137	6	6	374	1400	145	0.379	1.000	169	1.000	1.000	130	1.000	1.000	2946	2310559668	rs2238612	MAPP2	NM_002374.3:306A-G	synonymous	Unassigned	None	None	+	7
80	6	6	425	1400	64	0.594	1.000	55	0.902	0.970	59	1.000	1.000	2951	2311456637	rs1212781	CP12	NM_002212613.3:108A-G	missense	Unassigned	None	None	+	11
148	6	6	426	1400	42	0.931	1.000	57	0.902	1.000	59	1.000	1.000	2951	2311456639	rs1212553	CP12	NM_002212613.3:105C-T	synonymous	Unassigned	None	None	+	11
142	6	6	419	1400	129	0.419	1.000	292	1.000	1.000	297	1.000	1.000	3095	2310280163	rs1212608	CEP70	NM_024482.2:462A-G	synonymous	Unassigned	None	None	+	6
147	6	6	419	1400	129	0.419	1.000	292	1.000	1.000	297	1.000	1.000	3096	2310280122	rs1212607	CEP70	NM_024482.2:460A-A	missense	Unassigned	None	None	+	6
146	6	6	419	1400	129	0.419	1.000	292	1.000	1.000	297	1.000	1.000	3097	2310280862	rs1212612	FADA	NM_002398.3:439A-G	synonymous	Unassigned	None	None	+	2
66	6	6	479	1400	56	0.528	1.000	65	1.000	1.000	40	1.000	1.000	4027	3254818887	rs1212118	CHK36	NM_020865.2:1247C-G	missense	Unassigned	None	None	+	10
143	6	6	426	1400	42	0.463	1.000	95	1.000	1.000	40	1.000	1.000	4053	53193555	rs1212562	C1orf48	NM_001899342.3:184C-C	missense	Unassigned	None	None	+	2
153	6	6	449	1400	137	0.442	1.000	106	1.000	1.000	107	1.000	1.000	4066	53787873	rs1212133	MTRF	NM_002408.2:143A-G	missense	Unassigned	None	None	+	2
18	6	6	500	1400	16	0.375	1.000	15	1.000	1.000	23	1.000	1.000	4071	53278125	rs1212611	DNM5	NM_002369.2:1335A-G	synonymous	Unassigned	None	None	+	77
18	6	6	500	1400	16	0.420	0.930	24	1.000	1.000	26	1.000	1.000	4072	531931236	rs1212610	DNM5	NM_002369.2:1338A-G	missense	Unassigned	None	None	+	77
89	6	6	586	1400	49	0.512	1.000	79	1.000	1.000	97	1.000	1.000	5013	53353560	rs22554	ADAMTSL2	NM_030855.2:448C-C	missense	Unassigned	None	None	+	13
102	6	6	529	1400	137	0.474	1.000	127	1.000	1.000	109	1.000	1.000	5014	533549387	rs1212615	ADAMTSL2	NM_030855.2:4227C-T	synonymous	Unassigned	None	None	+	21
80	6	6	420	1400	10	0.460	1.000	43	1.000	1.000	41	1.000	1.000	5042	534584866	rs1212682	OPR12L	NM_024832.2:702C-G	missense	Unassigned	None	None	+	1
198	6	6	520	1400	240	0.422	1.000	255	0.957	1.000	203	0.961	1.000	5103	534914808	rs1212114	TSD4	NM_002429312.3:1832A-A	frameshift	Unassigned	None	None	+	2
15	6	6	333	1400	23	0.403	0.810	15	1.000	0.840	22	1.000	1.000	5124	534076232	rs1212613	TGCF1	NM_002302413.3:459C-T	missense	Unassigned	None	None	+	24
52	6	6	596	1400	38	0.530	1.000	307	1.000	1.000	40	1.000	1.000	5127	534912178	rs1212612	DNM5	NM_002369.2:1338A-G	missense	Unassigned	None	None	+	77
172	6	6	424	1400	150	0.440	1.000	146	0.993	1.000	138	1.000	1.000	5159	535082144	rs1212125	KAT2	NM_001442.2:5544C-T	synonymous	Unassigned	None	None	+	8
51	6	6	442	1400	52	0.421	1.000	57	1.000	1.000	56	1.000	1.000	5611	535999305	rs1212005	ATP2B8	NM_025153.2:432A-G	synonymous	Unassigned	None	None	+	26
14	6	6	442	1400	42	0.421	1.000	48	0.954	1.000	35	0.971	1.000	5612	535999374	rs1212006	ATP2B8	NM_025153.2:408A-C	synonymous	Unassigned	None	None	+	26
44	6	6	386	1400	79	0.443	1.000	67	1.000	1.000	65	1.000	1.000	5618	534072119	rs2229248	WDRB2	NM_021811.2:139C-T	synonymous	Unassigned	None	None	+	11
77	6	6	404	1400	56	0.446	1.000	68	0.995	1.000	40	1.000	1.000	5708	612085616	rs2256811	ELCV12	NM_017778.3:174C-T	synonymous	Unassigned	None	None	+	3
170	6	6	426	1400	36	0.633	1.000	133	1.000	1.000	67	0.948	1.000	5797	612191416	rs1212614	BAR1	NM_002398.3:419C-T	synonymous	Unassigned	None	None	+	4

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

as panel 'DLMP' with: All panel samples added between 5/17/2016 6/17/2016 Selected runs Selected samples OK

Regions of '800402.tvg-sorted_Output_Mutation_Report1_filtered'

Region Name	Chrom:Start - End	% Covered	Average Coverage	Minimum Coverage	Status	Average % Covered	Average Average Coverage	Average Minimum Coverage	Passed	Passed Percent	1
M_000251	2: 47630301 - 47630571	100%	280.93	91	Passed	100%	373.13	137.71	16	94.1176%	1
M_000251	2: 47635510 - 47635724	100%	433.34	247	Passed	100%	529.77	276.59	16	94.1176%	1
M_000251	2: 47637203 - 47637541	100%	451.48	129	Passed	100%	538.89	146.65	16	94.1176%	1
M_000251	2: 47639523 - 47639729	100%	445.34	266	Passed	100%	590.84	318.41	16	94.1176%	1
M_000251	2: 47641378 - 47641587	100%	372.54	93	Passed	100%	463.56	100.18	16	94.1176%	1
M_000251	2: 47643405 - 47643598	100%	569.91	319	Passed	100%	684.34	376.88	16	94.1176%	1
M_000251	2: 47656851 - 47657110	100%	574.01	296	Passed	100%	736.21	354.35	16	94.1176%	1

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.
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.

Table 7-2: Coverage Regions tab (Continued)

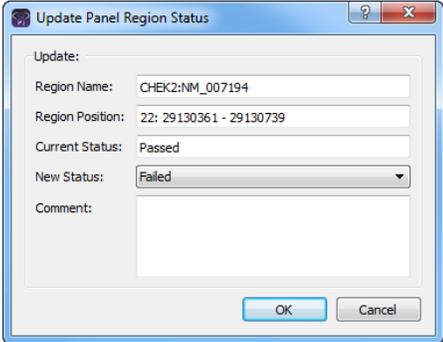
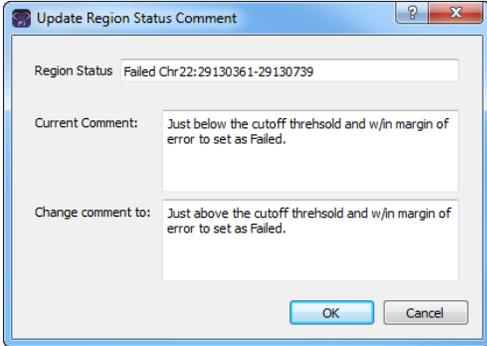
Column	Description
Minimum Coverage	The minimum coverage (number of reads aligned) for any position within the region.
% Covered Threshold	The percentage of the region that must have reads aligned to it, or the region does not pass the Quality Control settings that have been set for the run's panel. See "To specify the global Quality Control settings" on page 55.
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 55.
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 55.
Status	<p>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.</p> <p><i>Figure 7-14: Update Panel Region Status dialog box</i></p>  <p>Note: By default, only two statuses, Passed or Failed, are available, and one of these two statuses is always automatically selected for the region. You have the option of adding custom Panel Region statuses and manually selecting a status for a region. See "Managing Objects" on page 49.</p>

Table 7-2: Coverage Regions tab (Continued)

Column	Description
Status Comment	<p>Displays any comments that were added when the region status is manually changed. You can double-click in this field to add a new comment or modify an existing comment. After you add or edit a comment, and then click out of the field, the Update Region Status Comment dialog box opens.</p> <p><i>Figure 7-15: Update Status Region Comment dialog box</i></p>  <p>The dialog box displays the status and region positions for the selected region, the current (old) comment, and the new comment. You can make any additional edits to the new comment as needed, and then click OK to close the dialog box and add the new or modified comment for the region.</p>
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.
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 218.](#)) 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 “To add selected panel regions or variants to an Action List” on page 195.
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 267.
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 267.
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 37. 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 58.
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.
<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.

To add selected panel regions or variants to an Action List

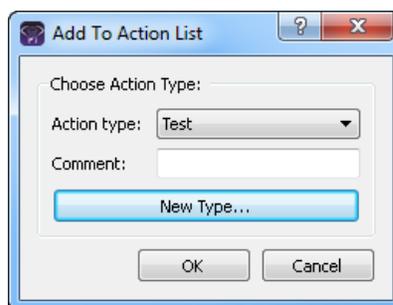
An *Action list* is a list of items that require some type of follow up. You can create multiple action lists relative to the type of follow up that is needed. You can create an Action List for panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab or for variants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

indicate that you want to review the coverage values for these regions.

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

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

Figure 7-16: Add To Action List dialog box



2. Click New Type.

The Add To Action List dialog box is refreshed with an option for adding an action type.
3. In the New Type field, enter an action type (for example, Review Coverage), and then click Add.

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

4. Repeat [Step 2](#) and [Step 3](#) as needed to add more action types.
5. On the Action Type dropdown list, select the appropriate action type.



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

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

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

Figure 7-17: Action List tab

ID	Region Name	Chrom	ChromRegion Start	ChromRegion End	Chrom:Start - End	% Covered	Average Coverage	Minimum Coverage	% C
154	CHEK2:NM_007194	22	29130361	29130739	22: 29130361 - 29130739	100%	148.87	90	0%
153	CHEK2:NM_007194	22	29121201	29121385	22: 29121201 - 29121385	100%	144.65	92	0%
152	CHEK2:NM_007194	22	29120935	29121142	22: 29120935 - 29121142	100%	188.45	91	0%



To save an Action List, you can generate a standard Action List Report. If multiple panes are included on the Action List tab, then by default, all these panes are included in the report. To remove a pane from the report, before generating the report, right-click the pane that you want to remove, and then click Remove. See [“Generating a Standard Report”](#) on page 269.

To calculate panel statistics (Run tab)

The Panel Statistics tab has options for [displaying](#) specific information based on a selected panel and panel group as well as any or all of the following criteria:

- All 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.
- Selected panel samples.

To display data on the Panel Statistics tab

1. If you have not already done so, open the Run tab for the appropriate run. (See [“Overview of the Run Tab”](#) on page 181.)
2. 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.



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.

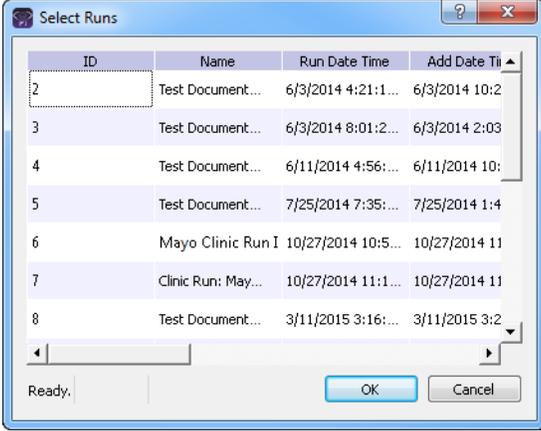
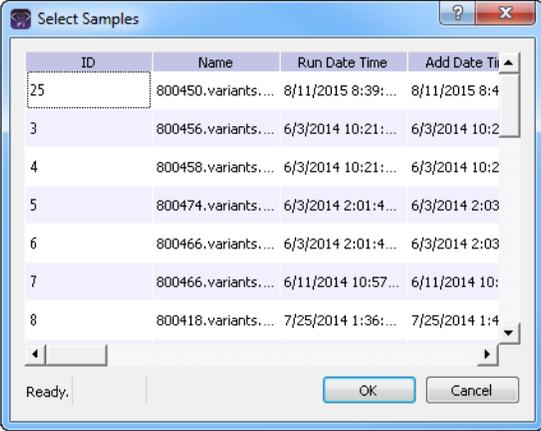
The Panel Statistics tab opens. The Coverage Regions sub-tab is the open tab. The panel and panel group that were used for the run are selected. You can select a different panel from the Panel dropdown list to view statistics for a different panel. You can also select a different panel to change the panel group, but you cannot change the panel group directly. By default, the statistics are calculated for all the samples in the selected run that have been added to the panel.

Figure 7-18: Coverage Regions sub-tab (Panel Statistics tab)

ID	Region Name	Chrom	ChronRegion Start	ChronRegion End	ChromStart - End	% Covered Threshold	Average Coverage Threshold	Minimum Coverage Threshold	Average % Covered	Average Average Coverage	Average M
154	CHEK2NM_007194	22	29130361	29130739	22:29130361-29130739	0%	0	0	127.445	80.5	
153	CHEK2NM_007194	22	29121201	29121385	22:29121201-29121385	0%	0	0	144.805	86.5	
152	CHEK2NM_007194	22	29120935	29121142	22:29120935-29121142	0%	0	0	182.535	83.5	
151	CHEK2NM_007194	22	29115353	29115503	22:29115353-29115503	0%	0	0	315.33	264.5	
150	CHEK2NM_007194	22	29107867	29108035	22:29107867-29108035	0%	0	0	412.125	305	
149	CHEK2NM_007194	22	29105964	29106077	22:29105964-29106077	0%	0	0	67.115	53.5	
148	CHEK2NM_007194	22	29099463	29099584	22:29099463-29099584	0%	0	0	258.845	221.5	
147	CHEK2NM_007194	22	29095796	29095955	22:29095796-29095955	0%	0	0	292.575	230	
146	CHEK2NM_007194	22	29092859	29093005	22:29092859-29093005	0%	0	0	332.56	229.5	
145	CHEK2NM_007194	22	29091668	29091891	22:29091668-29091891	0%	0	0	283.07	124	

3. Optionally, modify 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	<p>If you select this option, then by default, 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:</p> <ul style="list-style-type: none"> • 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																																
<p>Selected runs</p>	<p>To select specific runs for the panel, choose Selected runs, and then click the Browse button  that is next to Selected Runs to open the Select Runs dialog box. All the runs for the selected panel are displayed in the dialog box. Select the appropriate run, or CTRL-click to select multiple runs, and then click OK.</p> <p><i>Figure 7-19: Select Runs dialog box</i></p>  <table border="1" data-bbox="760 604 1261 913"> <thead> <tr> <th>ID</th> <th>Name</th> <th>Run Date Time</th> <th>Add Date Time</th> </tr> </thead> <tbody> <tr> <td>2</td> <td>Test Document...</td> <td>6/3/2014 4:21:1...</td> <td>6/3/2014 10:2</td> </tr> <tr> <td>3</td> <td>Test Document...</td> <td>6/3/2014 8:01:2...</td> <td>6/3/2014 2:03</td> </tr> <tr> <td>4</td> <td>Test Document...</td> <td>6/11/2014 4:56:...</td> <td>6/11/2014 10:</td> </tr> <tr> <td>5</td> <td>Test Document...</td> <td>7/25/2014 7:35:...</td> <td>7/25/2014 1:4</td> </tr> <tr> <td>6</td> <td>Mayo Clinic Run I</td> <td>10/27/2014 10:5:...</td> <td>10/27/2014 11</td> </tr> <tr> <td>7</td> <td>Clinic Run: May...</td> <td>10/27/2014 11:1:...</td> <td>10/27/2014 11</td> </tr> <tr> <td>8</td> <td>Test Document...</td> <td>3/11/2015 3:16:...</td> <td>3/11/2015 3:2</td> </tr> </tbody> </table>	ID	Name	Run Date Time	Add Date Time	2	Test Document...	6/3/2014 4:21:1...	6/3/2014 10:2	3	Test Document...	6/3/2014 8:01:2...	6/3/2014 2:03	4	Test Document...	6/11/2014 4:56:...	6/11/2014 10:	5	Test Document...	7/25/2014 7:35:...	7/25/2014 1:4	6	Mayo Clinic Run I	10/27/2014 10:5:...	10/27/2014 11	7	Clinic Run: May...	10/27/2014 11:1:...	10/27/2014 11	8	Test Document...	3/11/2015 3:16:...	3/11/2015 3:2
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7	Clinic Run: May...	10/27/2014 11:1:...	10/27/2014 11																														
8	Test Document...	3/11/2015 3:16:...	3/11/2015 3:2																														
<p>Selected samples</p>	<p>To select only specific samples for the panel, choose Selected samples and then click the Browse button  that is next to Selected samples to open the Select Samples dialog box. All the samples for the selected panel are displayed in the dialog box. Select the appropriate sample, or CTRL-click to select multiple samples, and then click OK.</p> <p><i>Figure 7-20: Select Samples dialog box</i></p>  <table border="1" data-bbox="760 1293 1261 1581"> <thead> <tr> <th>ID</th> <th>Name</th> <th>Run Date Time</th> <th>Add Date Time</th> </tr> </thead> <tbody> <tr> <td>25</td> <td>800450.variants....</td> <td>8/11/2015 8:39:...</td> <td>8/11/2015 8:4</td> </tr> <tr> <td>3</td> <td>800456.variants....</td> <td>6/3/2014 10:21:...</td> <td>6/3/2014 10:2</td> </tr> <tr> <td>4</td> <td>800458.variants....</td> <td>6/3/2014 10:21:...</td> <td>6/3/2014 10:2</td> </tr> <tr> <td>5</td> <td>800474.variants....</td> <td>6/3/2014 2:01:4...</td> <td>6/3/2014 2:03</td> </tr> <tr> <td>6</td> <td>800466.variants....</td> <td>6/3/2014 2:01:4...</td> <td>6/3/2014 2:03</td> </tr> <tr> <td>7</td> <td>800466.variants....</td> <td>6/11/2014 10:57...</td> <td>6/11/2014 10:</td> </tr> <tr> <td>8</td> <td>800418.variants....</td> <td>7/25/2014 1:36:...</td> <td>7/25/2014 1:4</td> </tr> </tbody> </table>	ID	Name	Run Date Time	Add Date Time	25	800450.variants....	8/11/2015 8:39:...	8/11/2015 8:4	3	800456.variants....	6/3/2014 10:21:...	6/3/2014 10:2	4	800458.variants....	6/3/2014 10:21:...	6/3/2014 10:2	5	800474.variants....	6/3/2014 2:01:4...	6/3/2014 2:03	6	800466.variants....	6/3/2014 2:01:4...	6/3/2014 2:03	7	800466.variants....	6/11/2014 10:57...	6/11/2014 10:	8	800418.variants....	7/25/2014 1:36:...	7/25/2014 1:4
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5	800474.variants....	6/3/2014 2:01:4...	6/3/2014 2:03																														
6	800466.variants....	6/3/2014 2:01:4...	6/3/2014 2:03																														
7	800466.variants....	6/11/2014 10:57...	6/11/2014 10:																														
8	800418.variants....	7/25/2014 1:36:...	7/25/2014 1:4																														

4. 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 200.](#)
 - Samples sub-tab. See [“Samples sub-tab \(Panel Statistics tab\)” on page 200.](#)
 - Variants sub-tab. See [“Variants sub-tab \(Panel Statistics tab\)” on page 201.](#)
 - Patients sub-tab. See [“Patients sub-tab \(Panel Statistics tab\)” on page 167.](#)
 - Reports sub-tab. See [“Reports sub-tab \(Panel Statistics tab\)” on page 168.](#)

Coverage Regions sub-tab (Panel Statistics tab)

The Coverage Regions sub-tab has a single pane, the Coverage Regions pane, that lists by default, the coverage information across all the samples of the selected run for every region in the selected panel and panel group. See [“Coverage Regions tab” on page 192](#) for a detailed discussion of the information that is displayed on this sub-tab.

Figure 7-21: Coverage Regions sub-tab (Panel Statistics tab)

ID	Region Name	Chrom	ChromRegion Start	ChromRegion End	Chrom:Start - End	% Covered Threshold	Average Coverage Threshold	Minimum Coverage Threshold	Average % Covered	Average Coverage	Average M
154	CHEK2-NM_007194	22	29130361	29130739	22:29130361-29130739	0%	0	0	100%	127.445	80.5
153	CHEK2-NM_007194	22	29121201	29121385	22:29121201-29121385	0%	0	0	100%	144.805	86.5
152	CHEK2-NM_007194	22	29120935	29121142	22:29120935-29121142	0%	0	0	100%	182.535	83.5
151	CHEK2-NM_007194	22	29115353	29115503	22:29115353-29115503	0%	0	0	100%	315.33	264.5
150	CHEK2-NM_007194	22	29107867	29108035	22:29107867-29108035	0%	0	0	100%	412.125	305
149	CHEK2-NM_007194	22	29105964	29106077	22:29105964-29106077	0%	0	0	100%	67.115	53.5
148	CHEK2-NM_007194	22	29099463	29099584	22:29099463-29099584	0%	0	0	100%	258.845	221.5
147	CHEK2-NM_007194	22	29095796	29095955	22:29095796-29095955	0%	0	0	100%	292.575	230
146	CHEK2-NM_007194	22	29092859	29093005	22:29092859-29093005	0%	0	0	100%	332.56	229.5
145	CHEK2-NM_007194	22	29091668	29091891	22:29091668-29091891	0%	0	0	100%	283.07	124

Samples sub-tab (Panel Statistics tab)

The Samples sub-tab has a single pane, the Samples pane, that lists by default, all the samples for the selected run that were imported for the selected panel and panel group. (See [Figure 7-22 on page 201.](#)) 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 215.](#)) 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 184.](#)

Figure 7-22: Panel Statistics tab, Samples pane

ID	Name	Run Date Time	Add Date Time	Run	Panel	Panel Group	Reference	Sample Groups	# Regions	# Regions Passed	Gt Content	Patient External ID	Status	Missed Clinical Variants
21	800402.variants.filter	4/7/2015 10:07:28 A...	4/7/2015 10:07:49 A...	Test Document...	DLMP	Default	Human_37_s92		154	0	N/A		N/A	C:\Users\Spec
20	800418.variants.filter	4/7/2015 9:42:44 AM	4/7/2015 9:49:20 AM	Test Document...	DLMP	Default	Human_37_s92		154	0	N/A		N/A	C:\Users\Spec
19	Sample_s3_738201_Ou...	4/7/2015 8:41:26 AM	4/7/2015 8:41:56 AM	Test Document...	DLMP	Default	Human_37_s92		154	0	N/A		N/A	C:\Users\Spec
18	Sample_s3_738201_Ou...	4/7/2015 8:41:26 AM	4/7/2015 8:41:55 AM	Test Document...	DLMP	Default	Human_37_s92		154	0	N/A		N/A	C:\Users\Spec
17	Sample_s2_799889_Ou...	4/7/2015 8:41:26 AM	4/7/2015 8:41:54 AM	Test Document...	DLMP	Default	Human_37_s92		154	0	N/A		N/A	C:\Users\Spec
16	800418.variants.filter	3/25/2015 10:59:07 ...	3/25/2015 11:05:35 ...	800402.variants...	DLMP	Default	Human_37_s92 Mayo Test 1		143	0	0		Review...	N/A
15	800402.variants.filter	3/25/2015 10:59:07 ...	3/25/2015 11:05:31 ...	800402.variants...	DLMP	Default	Human_37_s92 Mayo Test 1		143	0	0		New	N/A

Variants sub-tab (Panel Statistics tab)

The Variants sub-tab has a single pane, the Variants pane, that lists by default, all the variants that were detected in all the samples for the selected run that were imported 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 245.) 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 Sample Variant Data sub-tab on the Sample tab. See [“Sample Variant Data sub-tab”](#) on page 218.

Figure 7-23: Panel Statistics tab, Variants sub-tab

ID	Chromosome	Chromosome Position	Chr	ChrPos	Ref	Ref AA	Alt	Alt AA	Type	Coverage	Pathogenicity	Pathogenicity Status	Artifact Type	Gene	Gene Strand	Exon Number	Transcript	Protein
50	2	48033891	2	48033891	r1587773207	-	T		131		Unassigned	None	Sequencing	MSH4	-	intron	NM_000129.2	NP_000170.1
49	2	48033891	2	48033891	r1207608137	T	-		195		Unassigned	None	Sequencing	MSH4	-	intron	NM_000129.2	NP_000170.1
48	22	29130458	22	29130458	r11805129	T	C		99		Unassigned	None	Sequencing	CHEK2	-	5' UTR	NM_001257387.1	NP_001244316.1
47	2	47693941	2	47693941	C	Att	-	Thr57Ser4	frame-shift	370		Unassigned	None	MSH4	-	11	NM_001259281.1	NP_001245210.1
46	18	48584856	18	48584856	r1286387076	-	TT		223		Unassigned	None	Sequencing	SMAAD4	-	intron	NM_005559.5	NP_005550.1
45	10	89720634	10	89720634	T	-			241		Unassigned	None	Sequencing	PTEF1	-	intron	NM_000314.4	NP_000305.3
42	10	88683122	10	88683122	r12074064	T	C		289		Unassigned	None	Sequencing	BMP2L1A	-	intron	NM_004320.2	NP_004320.2
41	10	88635779	10	88635779	r11528010	C	Pro	A Thr	missense	547		Unassigned	None	BMP2L1A	-	3	NM_004320.2	NP_004320.2
37	2	47693788	2	47693788	r11299837	A	T		317		Unassigned	None	Sequencing	MSH4	-	intron	NM_001259281.1	NP_001245210.1
30	19	1222012	19	1222012	r12073607	G	C		171		Unassigned	None	Sequencing	STK11	-	intron	NM_000455.4	NP_000446.1

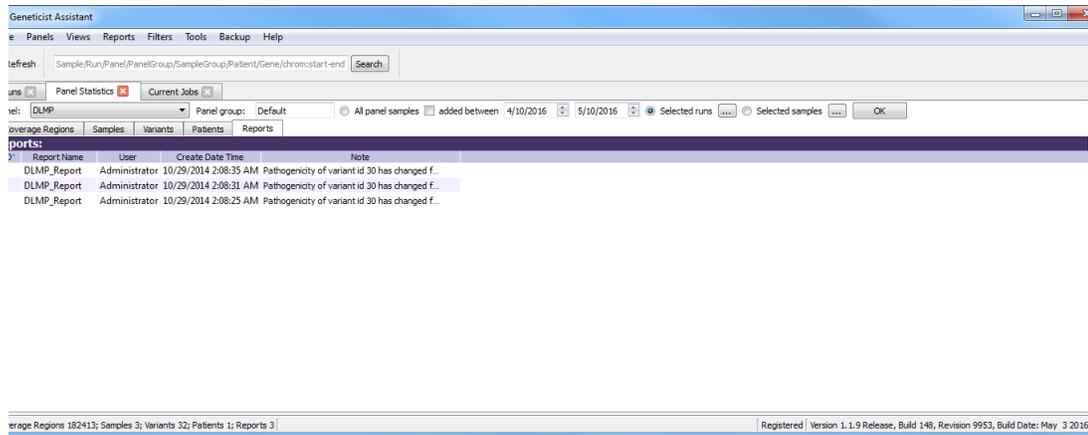
Reports sub-tab (Panel Statistics tab)

The Reports sub-tab contains a single pane, the Reports pane, that lists by default, all the reports that were generated for the samples for the selected run that were imported for the selected panel and panel group and saved to the Geneticist Assistant database. See [Figure 7-24](#) on page 202.



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

Figure 7-24: 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 126](#).



For detailed information about generating and saving Panel Statistics reports, see [Chapter 10, “Geneticist Assistant Reports,” on page 267](#).

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



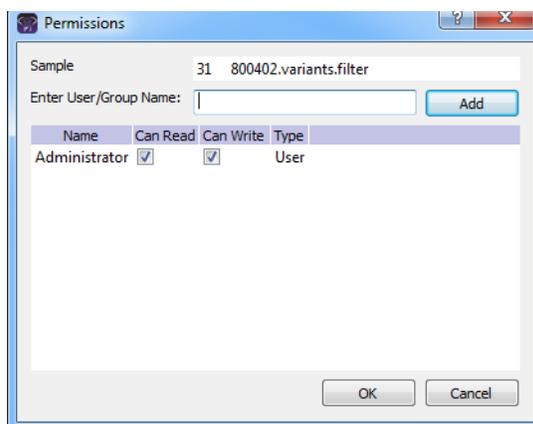
You can assign Read and Write permissions for all the samples in a run in a single step. See [“To set or modify the Read/Write permissions for a run” on page 168](#).

1. If you have not already done so, open the Run tab. (See [“Overview of the Run Tab” on page 181](#).)
2. In the Samples pane, select the samples, or CTRL-click to select the multiple samples for which the permissions are being set.
3. Right-click any selected sample, and on the context menu that opens, click Permissions.

The Permissions dialog box opens. See [Figure 7-25 on page 203](#).

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

Figure 7-25: Permissions dialog box (Run tab, Samples pane)



4. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to [Step 5](#).



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.

5. 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.



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.

6. Select the appropriate 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 Read permission is already assigned for the selected user or group. If just this permission is selected, then the user or group can simply view the sample record. They cannot modify it.
7. Optionally, if the user or group must be able to modify the sample record as well, then select the Can Write permission.
8. Click OK.
A Permission Updated message opens.

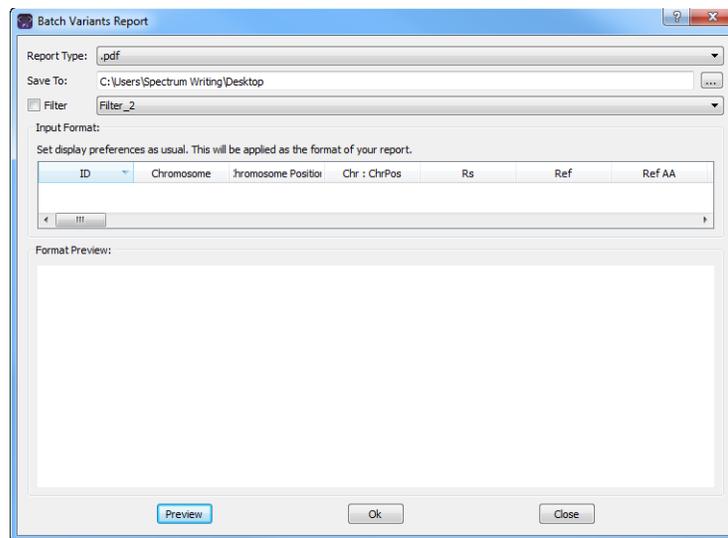
9. Click OK.

Both the message and the Permissions dialog box close. The specified permissions are set for the sample record.

To manage a Batch Variants report (Run tab)

A *Batch Variants report* is a report that includes information for variants in a selected sample, and allows you to create multiple reports for multiple samples in a single step. If you generate a Batch Variants report from the Run tab, then a separate report is saved for each of the selected samples in the run. A *Batch Variants report configuration* determines the data that is to be included 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 (PDF, HTML and so on), or the location in which the report is to be saved. Instead, you must specify the format and location in which to save the report every time you generate a report for a new or saved configuration.

Figure 7-26: *Batch Variants Report dialog box*



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

- [“To generate a Batch Variants report based on a new configuration”](#) below.
- [“To generate a Batch Variants report based on a saved configuration”](#) on page 206.
- [“To modify a Batch Variants report configuration”](#) on page 207.
- [“To delete a Batch Variants report configuration”](#) on page 208.



To generate the report for all variants in all the samples for the run, see “To manage a Batch Variants report (Runs tab)” on page 170.)

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 the first sample in the list of samples 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.

1. If you have not already done so, open the Run tab.
2. In the Samples pane, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, on the Report Type dropdown list, select the report type. (The default value is PDF.)
5. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
 - Leave this location as-is, and then go to [Step 6](#).
 - 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.
6. Optionally, to filter the data that is to be displayed in the report, click 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](#).

7. Optionally, to set the display preferences for the report, do any of the following as needed:
 - 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 each of the selected samples. After the report is successfully generated, a Batch Variants Report Finished dialog box with a message that indicates that the report was generated and where the report was saved.

10. Click OK.

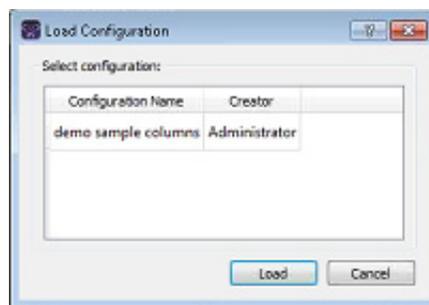
The dialog box closes. You return to a blank Current Jobs tab on the Geneticist Assistant main window.

To generate a Batch Variants report based on a saved configuration

1. If you have not already done so, open the Run tab.
2. In the Samples pane, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.
4. On the Batch Variants Report dialog box, select the output format for the report.
5. If a Batch Variants report has previously been generated and saved, then the location in which the report was saved is displayed in the Save To field when the Batch Variants Report dialog box opens; otherwise, this field is blank. Do one of the following:
 - Leave the location as-is, and then go to [Step 6](#).
 - 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.
6. Right-click on any heading in the Input Format pane, and then choose Select Columns. The Select Columns dialog box opens.
7. 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 7-27: Load Configuration dialog box



8. Select a configuration, and then click Load.

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

9. Click Close (x) to close the Select Columns dialog box.
10. Optionally, to preview an onscreen preview of the report before it is generated and saved, click Preview.
11. Click OK.

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

12. Click OK.

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

To modify a Batch Variants report configuration

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

1. If you have not already done so, open the Run tab.
2. In the Samples pane, select the sample or CTRL-click to select the multiple samples for which the Batch Variant report is to be generated.
3. Right-click any selected sample, and on the context menu that opens, click Export > Create Batch Variants report.

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

4. 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. To modify the report configuration, see [“To create/modify a pane configuration” on page 39](#).

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

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

9. 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. On the Batch Variants Report dialog box, right-click on any heading in the Input Format pane, and then choose Select Columns.
The Select Columns dialog box opens.
2. See [“To delete a pane configuration” on page 41](#).
3. Click Close (x) to close the Batch Variants Report dialog box.

To generate a Batch Coverage report (Run tab)

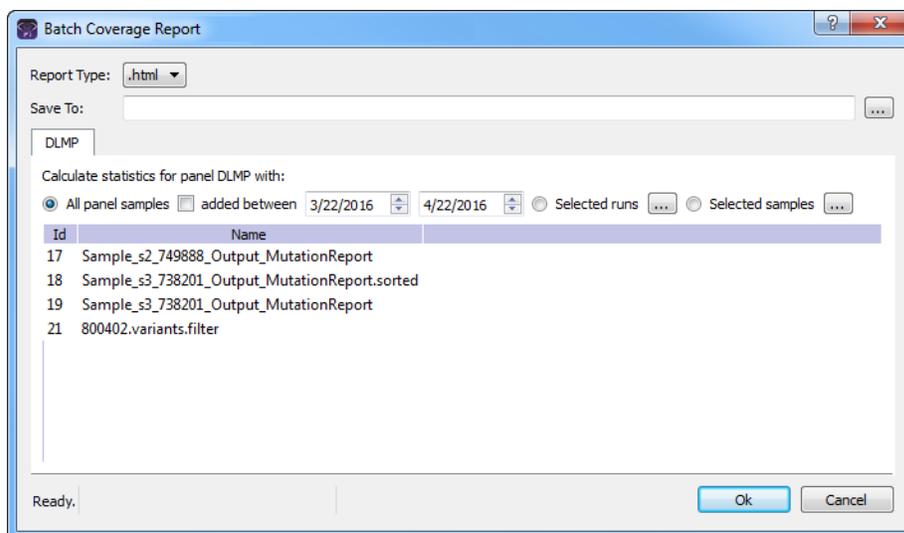
The Batch Coverage report generates a coverage report for each of the selected samples in a run in a specific panel or panels and panel group. The report also includes coverage statistics for the panel that are calculated based on one of the following options:

- All 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.
 - Selected panel samples.
1. If you have not already done so, open the Run tab.

- In the Samples pane, right-click a sample (CTRL-click to select multiple samples), and on the context menu that opens, click Export > Create Batch Coverage report.

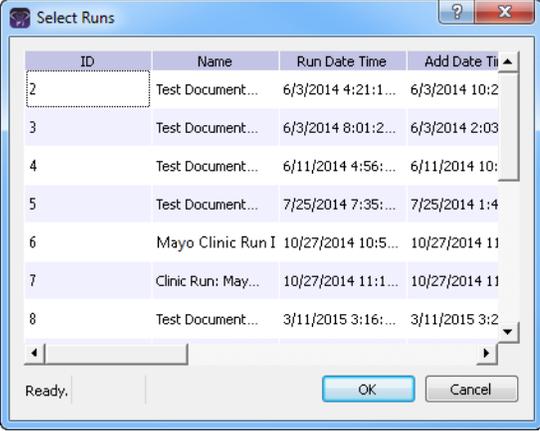
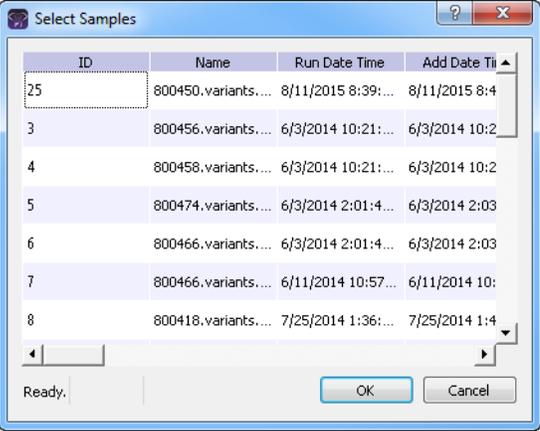
The Batch Coverage Report dialog box opens. All the selected samples for the run are displayed in the bottom pane of the dialog box. If multiple samples are selected with different panels, then a separate tab is displayed for each panel.

Figure 7-28: Batch Coverage Report dialog box (Runs tab)



- Leave the report type set to the default value of HTML, or select a different report type.
- 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.
- To calculate the panel coverage statistics for the report based on all panel samples, click OK; otherwise, specify the different options by which to calculate the panel coverage statistics, and then click OK.

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: <ul style="list-style-type: none"> 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
<p>Selected runs</p>	<p>To select specific runs for calculating the panel coverage statistics, choose Selected runs, and then click the Browse button  that is next to Selected runs to open the Select Runs dialog box. All the runs for the panel are displayed in the dialog box. Select the appropriate run, or CTRL-click to select multiple runs, and then click OK.</p> <p><i>Figure 7-29: Select Runs dialog box</i></p> 
<p>Selected samples</p>	<p>To select only specific samples for calculating the panel coverage statistics, choose Selected samples and then click the Browse button  that is next to Selected samples to open the Select Samples dialog box. All the samples for the panel are displayed in the dialog box. Select the appropriate sample, or CTRL-click to select multiple samples, and then click OK.</p> <p><i>Figure 7-30: Select Samples dialog box</i></p> 



If the panel coverage statistics are to be calculated 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 Runs tab.

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 for each selected sample in the specified format and the specified location.

Run's Status Changes pane

The Run's Status Changes pane shows the history of all the status changes that have been made for the run in chronological order. A New status entry is always displayed for a run. You can right-click anywhere on this pane to open a context menu with 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 267.
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 37.</p> <p>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 58.</p>
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 126.



For detailed information about generating and saving both standard and custom Run reports, see [Chapter 10, “Geneticist Assistant Reports,”](#) on page 267.

Chapter 7
Geneticist Assistant Data Review - Run Tab

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” on page 217.](#)
- [“Sample Variant Data sub-tab” on page 218.](#)
- [“Sample Data sub-tab” on page 238.](#)

Chapter 8
Geneticist Assistant Data Review - Sample Tab

Overview of the Sample Tab

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

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

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

ID	Chromosome	Chromosome Position	Chr : ChrPos	Rs	Ref	Ref AA	Alt	Alt AA	Type	Coverage	Pathogenicity	Pathogenicity Status	Artifact Type
48	22	29130458	22:29130458	rs1805129	A		G		synonymous	99	Unassigned		None
47	2	47693941	2:47693941		C	Asn	-	Thr	frame-shift	376	Likely Deleterious		None
46	18	48584856	18:48584856	rs57847829	-			TT		299	Unassigned		None
45	10	89720634	10:89720634		T		-			258	Benign		None
44	2	48033891	2:48033891	rs267608137	T		-			254	Unassigned	Pending	None
42	10	88683122	10:88683122	rs7074064	T		-			289	Unassigned		None
41	10	88635779	10:88635779	rs11528010	C	Pro	A	Thr	missense	548	Unassigned		None
37	2	47693788	2:47693788	rs12998837	A		T			317	Unassigned		None
30	19	1222012	19:1222012	rs2075607	G		C			171	Unassigned		None
21	2	47693959	2:47693959	rs3732183	G		A			302	Unassigned		None
18	17	63554591	17:63554591	rs2240308	C	Pro	T	Ser	missense	522	Unassigned		None
17	17	63533789	17:63533789	rs9915936	A		G		synonymous	264	Unassigned		None
16	17	63533768	17:63533768	rs1132683	C		T		synonymous	252	Unassigned		None
15	17	7579472	17:7579472	rs1042322	C		G			349	Unassigned		None
14	14	75513883	14:75513883	rs175081	A	Asn	G	Asp	missense	356	Unassigned		None

The Sample tab has two sub-tabs:

- The Sample Variant Data sub-tab. See “Sample Variant Data sub-tab” on page 218.
- The Sample Data sub-tab. See “Sample Data sub-tab” on page 238.

Sample Variant Data sub-tab

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

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

ID	Chromosome	Chromosome Position	Chr : ChrPos	Rs	Ref	RefAA	Alt	Alt AA	Type	Coverage	Pathogenicity	Pathogenicity Status	Artifact Type
48	22	29130458	22:29130458	rs1805129	A	G			synonymous	99	Unassigned		None
47	2	47693941	2:47693941		C	Asn	-	Thr	frame-shift	376	Likely Deleterious		None
46	18	48584856	18:48584856	rs57847829	-				TT	299	Unassigned		None
45	10	89720634	10:89720634		T					258	Benign		None
44	2	48033891	2:48033891	rs267608137	T					254	Unassigned	Pending	None
42	10	88683122	10:88683122	rs7074064	T	C				289	Unassigned		None
41	10	88635779	10:88635779	rs11528010	C	Pro	A	Thr	missense	548	Unassigned		None
37	2	47693788	2:47693788	rs12998837	A	T				317	Unassigned		None
30	19	1222012	19:1222012	rs2075607	G	C				171	Unassigned		None
21	2	47693959	2:47693959	rs3732183	G	A				302	Unassigned		None
18	17	63554591	17:63554591	rs2249308	C	Pro	T	Ser	missense	522	Unassigned		None
17	17	63533789	17:63533789	rs9915936	A	G			synonymous	264	Unassigned		None
16	17	63533768	17:63533768	rs1133683	C	T			synonymous	252	Unassigned		None
15	17	7579472	17:7579472	rs1042522	C	G				349	Unassigned		None
14	14	75513883	14:75513883	rs175081	A	Asn	G	Asp	missense	356	Unassigned		None

The Sample Variant Data sub-tab has a single pane, the Variants pane, that displays information about all the variants that were identified in the selected sample. By default, two types of information are 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 219.
- “Global variant count and frequency information” on page 224.
- “Sample group statistical information” on page 225.
- “Variant options on the Sample Variant Data sub-tab” on page 226.
- “Sample Variant Data sub-tab context menu” on page 228.



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



- Remember, you can select which columns to show and hide in a pane. See “To show/hide columns in a tab pane” on page 37.
- Remember, you can specify how the information about the variants is displayed on the sub-tab. See “To specify variant preferences” on page 60.
- 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 245.

Variant annotation information

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 chromosome: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)

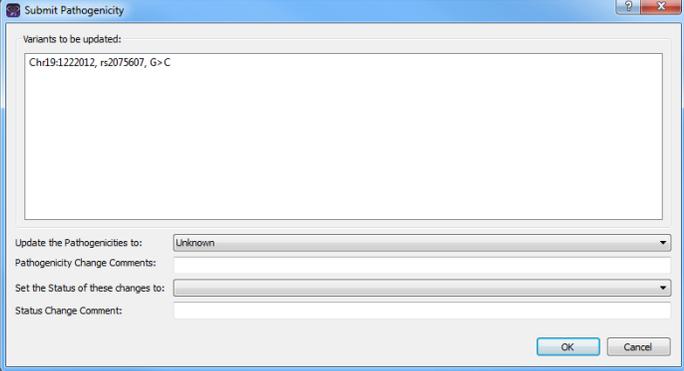
Column	Description
Pathogenicity	<p>The pathogenicity of the variant, which is the potential for the variant to cause disease. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be; for example, Deleterious. (If you cannot determine the pathogenicity of the variant even after reviewing all the relevant information, you can set the status to Unknown.) After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant. To open the Submit Pathogenicity Status dialog box, double-click in this column for the selected variant. You can update the pathogenicity for the selected variant and/or the status of the pathogenicity in this dialog box. You can also add a comment about the pathogenicity change and/or pathogenicity status change</p> <p>Note: The option to update the pathogenicity and/or pathogenicity status (“Update Pathogenicity”) is also available on the context menu for the sub-tab.</p> <p><i>Figure 8-3: Submit Pathogenicity dialog box</i></p>  <p>The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant:</p> <ul style="list-style-type: none"> • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See “Managing Objects” on page 49. • Optionally, you can add supporting information for this pathogenicity change. See “To add supporting information for a variant” on page 257. • You always have the option of updating just the pathogenicity status for a variant at a later date. See “Pathogenicity Status” below.

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

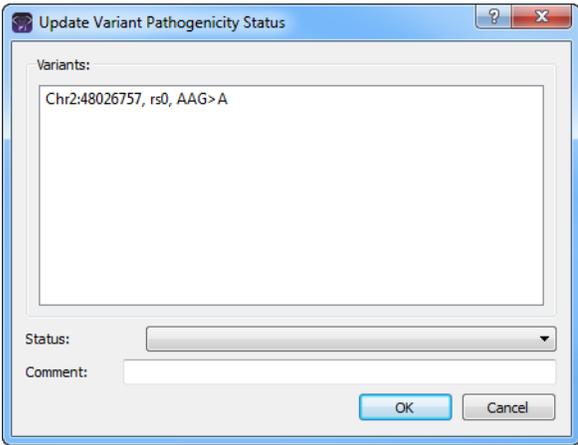
Column	Description
Pathogenicity Status	<p>The status of the selected pathogenicity. You must manually set this value. To open the Update Variant Pathogenicity Status dialog box, double-click in this column for the selected variant. You can set just the pathogenicity status for the variant in this dialog box. You can also add a comment about the status change.</p> <p>Note: The option to update just the status (“Update Pathogenicity Status”) is also available on the context menu for the sub-tab.</p> <p><i>Figure 8-4: Update Variant Pathogenicity Status dialog box</i></p> 

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

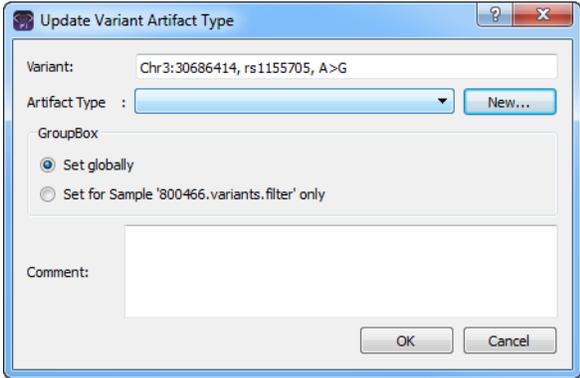
Column	Description
Artifact Type	<p>The artifact type for the selected variant. You must manually set this value. Double-click in this column for the selected variant to open the Update Artifact Type dialog box. You specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type.</p> <p>Note: The option to update the artifact type (“Update Artifact Type”) is also available on the context menu for the sub-tab.</p> <p><i>Figure 8-5: Update Variant Artifact Type dialog box</i></p>  <p>When you specify the artifact type, you can set the artifact type as the following:</p> <ul style="list-style-type: none"> • 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. <p>You also have the option of adding comments for the variant artifact type. The following information is relevant to updating the artifact type for a variant:</p> <ul style="list-style-type: none"> • You can CTRL-click to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant on the Variants sub-tab on the Patient tab, on the Variants sub-tab on the Panel Statistics tab from the Runs tab or the Run tab, or on the Sample Variant Data sub-tab on the Sample tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See “Managing Objects” on page 49.
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.
Zygotity	Indicates whether the variant is heterozygous or homozygous. Note: The selected zygotity is based on the Zygotity settings that you have specified on the Variant Preferences tab of the Settings dialog box. See “To specify variant preferences” on page 60 .
Read Balance	The balance in forward and reverse oriented reads with the variant as reported in the VCF file.
Gene	The gene on which the variant is located.
Gene Strand	The strand (+ or -) for the gene in which the variant is located.
Exon Number	The exon within the gene in which the variant is located. “Intron” is displayed for intronic variants.

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

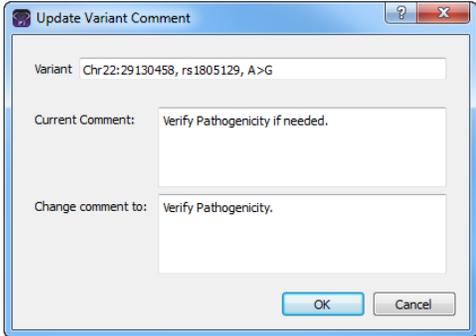
Column	Description
Transcript	The accession number for the preferred mRNA transcript that is used for the HGVS nomenclature columns.
Protein	The accession number for the protein transcript that is associated with the preferred mRNA transcript and is used for the HGVS nomenclature columns.
Coding Base	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	<p>The last added comment for the variant. You can also double-click in this field, and then add or edit a comment as needed. After you click out of the field, the Update Variant Comment dialog box opens.</p> <p><i>Figure 8-6: Update Variant Comment dialog box</i></p>  <p>The dialog box displays the variant for which you are adding or editing the comment, the current (old) comment, and the new comment. You can make additional edits to the new comment needed, and then click OK to close the dialog box and save the new or modified comment for the variant.</p> <p>Note: 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 229.</p> <p>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.</p>
Times Observed Per Run	The number of times that the variant was detected across all samples in the run.
Panel	The panel that was used for the run.

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

Column	Description
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.
HGMD Mutation	The amino acid changes that was caused by the variant as recorded in HGMD.
HGMD HGVS	The HGVS code relative to HGMD.
HGMD ID	The ID for the variant in HGMD.
Note: If you have not specified your login settings for the HGMD database (see “To specify HGMD settings” on page 62), then these last three columns are not displayed.	

Global variant count and frequency information

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

Table 8-2: Sample Variant Data sub-tab: Global Variant Count and Frequency information (Continued)

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

Sample group statistical information



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 228](#).

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.

Table 8-3: Sample Group Statistics Columns on the Sample Variant Data sub-tab (Continued)

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

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:

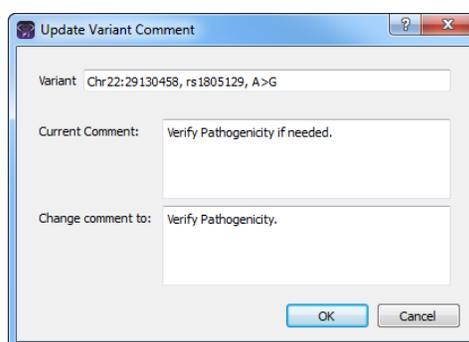
- You can specify how the information about the variants is to be displayed on this sub-tab. See [“To specify variant preferences” on page 60](#).
- If the variant is located where there are overlapping genes, then you can select a different gene for the variant on the Gene dropdown list. If you select 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 92](#).
- 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 [“Sample Variant Data sub-tab context menu”](#) on page 228), 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 90.

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

Figure 8-7: Update Variant Comment dialog box



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



You can also use the *Add Variant Comment* option that is available on the context menu for the variant to add a new variant comment. See [“Add Variant Comment”](#) on page 229.

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

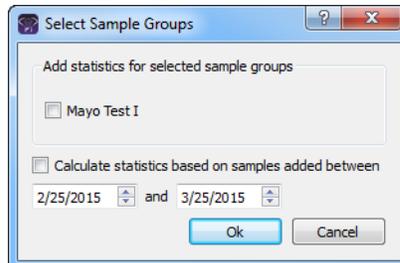


If you make a change to any of these values for the variant, then these changes are recorded for the variant on the *Variant Changes* pane on the *Variant* tab. See [“Variant Changes pane”](#) on page 253. These changes are also reflected in the appropriate fields on all the variant tabs, sub-tabs, and panes in the Geneticist Assistant application.

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 8-8: Select Sample Groups dialog box



- If you used sample groups for a run, then you can indicate the date range for which to calculate statistics based on the added samples.
- If you did not use sample groups for a run, you can clear the options for selected sample groups, which hides the corresponding Sample Group Statistics columns from the sub-tab display.

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 245.
Show Variants Filtered by Panel	Displays all the variants in the selected sample filtered by a selected panel. If you select this option, then the Select Panel to Filter Variants dialog box opens. You must select a panel in this dialog box by which to filter the variants for the selected run. <i>Figure 8-9: Select Panel to Filter Variants dialog box</i>
Show Samples with Selected Variants	Opens the Samples with variants pane, which displays a list of all the samples in your Geneticist Assistant database that contain the selected variant. 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 184.

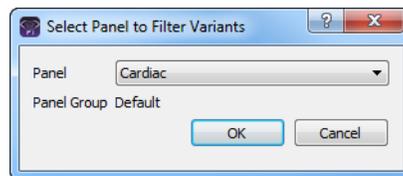


Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

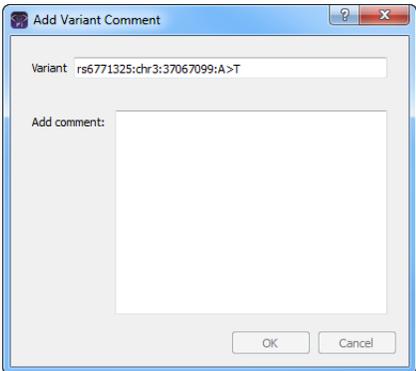
Option	Description
Edit Variant - Contains all the sub-menu options for editing and deleting a variant.	
Add Variant Comment	<p>Opens the Add Variant Comment dialog box. You can add a comment for the variant in the Add Comment field, and then click OK. You can also CTRL-click to select multiple variants 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.</p> <p><i>Figure 8-10: Add Variant Comment dialog box</i></p>  <p>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.</p>

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

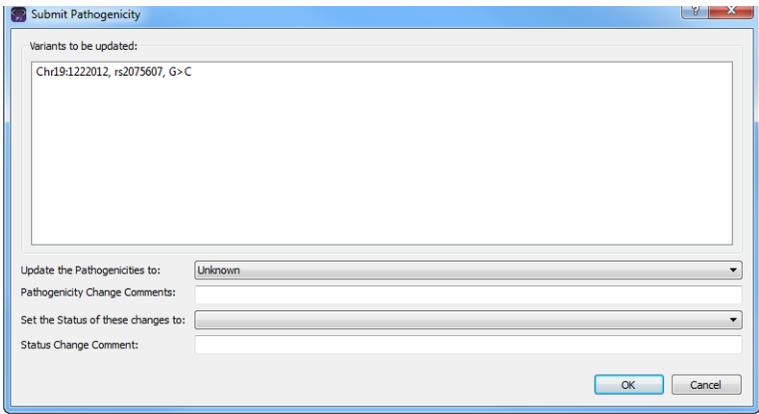
Option	Description
Update Pathogenicity	<p>Opens the Submit Pathogenicity dialog box. You can update the pathogenicity for the selected variant and/or the status of the pathogenicity in this dialog box. You can also add a comment about the pathogenicity change and/or pathogenicity status change. By default, the pathogenicity is always set to an initial value of Unassigned. You must review the information for the variant and then decide what the pathogenicity should be; for example, Deleterious. (If you cannot determine the pathogenicity of the variant even after reviewing all the relevant information, you can set the status to Unknown.) After you set the pathogenicity for a variant, the next time that the variant is detected in another sample, this is the pathogenicity that is displayed for the variant.</p> <p>Figure 8-11: Submit Pathogenicity dialog box</p>  <p>Note: You can also double-click in the Pathogenicity column to open the Submit Pathogenicity dialog box. You can also CTRL-click to select multiple variants for updating the pathogenicities and the pathogenicity statuses at the same time.</p> <p>Note: You can also update the Pathogenicity and the Pathogenicity Status for a variant on the Variants sub-tab on the Patient tab.</p> <p>The following information is relevant to updating the pathogenicity and/or pathogenicity status for a variant:</p> <ul style="list-style-type: none"> • By default, only five statuses—Deleterious, Likely Deleterious, Unknown, Likely Benign, and Benign—are available. You have the option of adding custom pathogenicities. See “Managing Objects” on page 49. • Optionally, you can add supporting information for this pathogenicity change. See “To add supporting information for a variant” on page 257. • You always have the option of updating just the pathogenicity status for a variant at a later date. See “Update Pathogenicity Status” below.

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

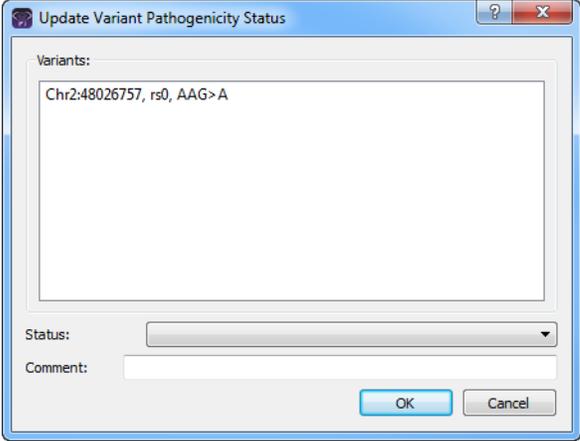
Option	Description
Update Pathogenicity Status	<p>Opens the Update Variant Pathogenicity Status dialog box. You can update the pathogenicity status for the variant in this dialog box. You can also add a comment for the variant.</p> <p><i>Figure 8-12: Update Variant Pathogenicity Status dialog box</i></p>  <p>Note: By default, only two statuses, Pending or Confirmed, are available. You have the option of adding custom pathogenicity statuses. See “Managing Objects” on page 49.</p> <p>Note: You can also update the Pathogenicity Status for a variant on the Variants sub-tab on the Patient tab.</p> <p>Note: You can also double-click in the Pathogenicity Status column for a variant to open the Update Variant Pathogenicity Status dialog box. You can also CTRL-click to select multiple variants for updating the pathogenicity statuses at the same time.</p>

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

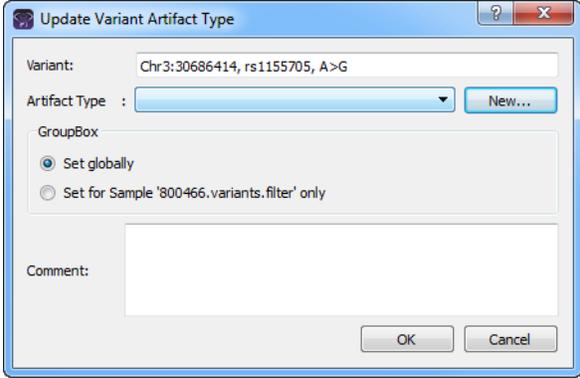
Option	Description
Update Artifact Type	<p>Opens the Update Artifact Type dialog box. You can specify the artifact type (for example, Sequencing) for the variant in this dialog box. If the appropriate artifact type is not available, then an option is provided to create a new type.</p> <p><i>Figure 8-13: Update Variant Artifact Type dialog box</i></p>  <p>When you specify the artifact type, you can set the artifact type as the following:</p> <ul style="list-style-type: none"> • 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. <p>You also have the option of adding comments for the variant artifact type. The following information is relevant to updating the artifact type for a variant:</p> <ul style="list-style-type: none"> • You can CTRL-click to select multiple variants for updating the artifact type at the same time. • You can also define artifact types and set the artifact type for a variant on the Variants sub-tab on the Patient tab, or on the Variants sub-tab on the Panel Statistics tab from the Runs or the Run tab. • By default, only a single artifact type, None, is available. You have the option of adding custom artifact types. See “Managing Objects” on page 49.

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

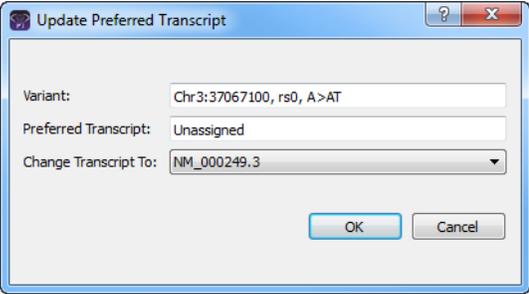
Option	Description
Update Preferred Transcript	<p>Opens the Update Preferred Transcript dialog box. You can select a different preferred transcript for the variant on the Change Transcript To dropdown list.</p> <p><i>Figure 8-14: Update Preferred Transcript dialog box</i></p>  <p>Note: When you select a different transcript here, 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 90. 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.</p>
Delete Variant	Deletes the selected variant from the sample.
External - Contains all the sub-menu options for viewing the variant in third-party tools.	
View in Alamut	<p>Opens the variant for viewing in Alamut.</p> <p>Note: To view a variant in Alamut, your Alamut settings must be specified. See “To specify Alamut settings” on page 61.</p>
View in UCSC	Opens the variant for viewing in the UCSC Variant Genome Browser.

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

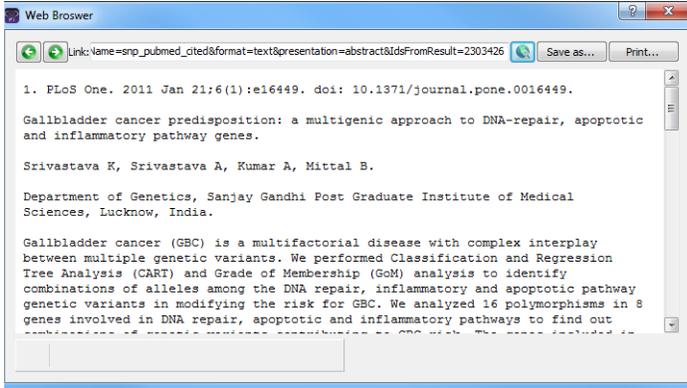
Option	Description
View PubMed Abstract	<p>Searches PubMed for the variant and displays the appropriate abstract in the PubMed window.</p> <p><i>Figure 8-15: PubMed window</i></p>  <p>You can do the following for the abstract:</p> <ul style="list-style-type: none"> • In the Link field, enter a different 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 “To add selected panel regions or variants to an Action list” on page 235.
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 267.
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 267.

Table 8-4: Sample tab, Sample Variant Data sub-tab context menu (Continued)

Option	Description
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 37.</p> <p>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 58.</p>
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.

To add selected panel regions or variants to an Action list

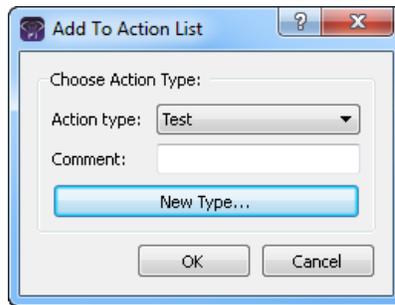
An *Action list* is a list of items that require some type of follow up. You can create multiple action lists relative to the type of follow up that is needed. You can create an Action List for panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab or for variants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

1. Do one of the following:
 - To select a single panel region for the Action list, right-click the appropriate panel region on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab, and on the context menu that opens, click Add Selected Regions to Action List.
 - To select multiple panel regions, press and hold the CTRL key as you select the appropriate panel regions on the Coverage Regions tab or the Coverage Regions sub-tab on the Coverage QC tab, and then right-click any selected panel region, and on the context menu that opens, click Add Selected Regions to Action List.

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

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

Figure 8-16: Add To Action List dialog box



2. Click New Type.

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

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

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

4. Repeat [Step 2](#) and [Step 3](#) as needed to add more action types.
5. On the Action Type dropdown list, select the appropriate action type.



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

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

7. Click OK.

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

Figure 8-17: Action List tab

ID	Region Name	Chrom	ChromRegion Start	ChromRegion End	Chrom:Start - End	% Covered	Average Coverage	Minimum Coverage	% C
154	CHEK2:NM_007194	22	29130361	29130739	22: 29130361 - 29130739	100%	148.87	90	0%
153	CHEK2:NM_007194	22	29121201	29121385	22: 29121201 - 29121385	100%	144.65	92	0%
152	CHEK2:NM_007194	22	29120935	29121142	22: 29120935 - 29121142	100%	188.45	91	0%

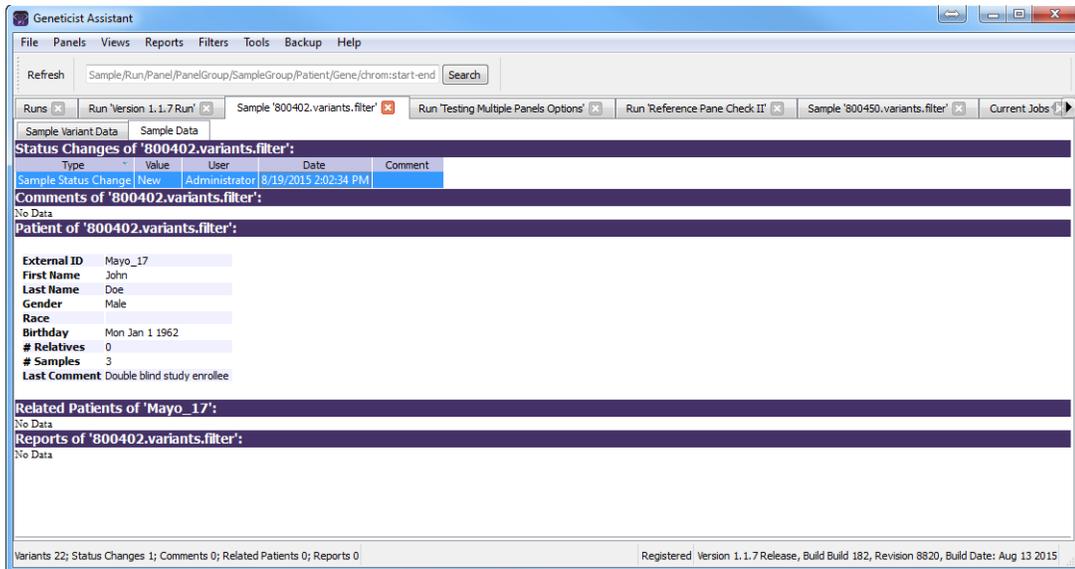


To save an Action List, you can generate a standard Action List Report. If multiple panes are included on the Action List tab, then by default, all these panes are included in the report. To remove a pane from the report, before generating the report, right-click the pane that you want to remove, and then click Remove. See “Generating a Standard Report” on page 269.

Sample Data sub-tab

The Sample Data sub-tab contains five panes—Status Changes, Comments, Patient, Related Patients, and Reports—that provide different information about the sample.

Figure 8-18: Sample tab, Sample Data sub-tab



A context menu is available for each pane. See:

- “Status Changes pane” below.
- “Comments pane (Sample Data sub-tab on the Sample tab)” on page 240.
- “Patient pane (Sample Data sub-tab for the Sample tab)” on page 241.
- “Related Patients pane” on page 242.
- “Reports pane (Sample data sub-tab on the Sample tab)” on page 244.

Status Changes pane

The Status Changes pane lists an entry for each time the status was changed for the sample. Each entry details the value for the sample status, the name of the user who changed the sample status, the date and time of the status change, and any comments that the user entered in support of the change. The pane always contains an entry for the initial submission of the sample with the status of the sample set to New.



Remember, you can change the status for a sample on the Run tab. See “To set the sample status” on page 182.

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 267 .
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 37.</p> <p>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 58.</p>
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. You can-right click a comment in the pane to open a context menu with the following options:

Table 8-6: Sample tab, Sample Data sub-tab, Comments pane context menu

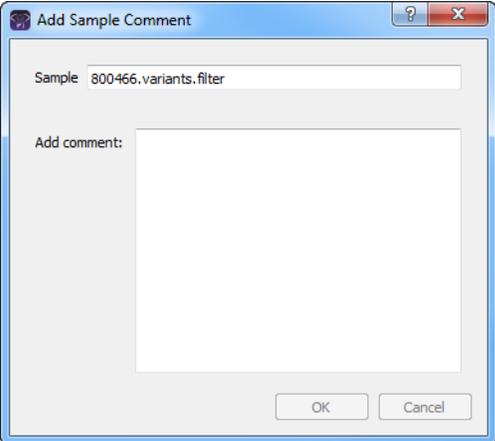
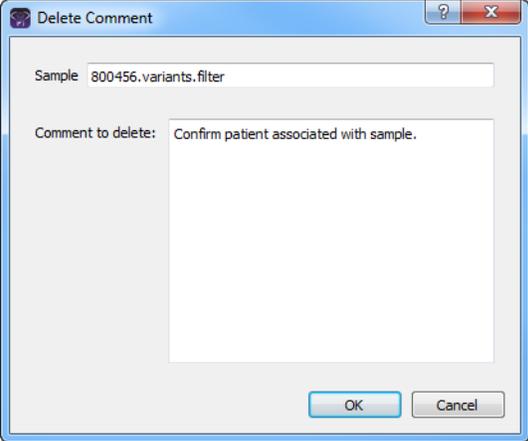
Option	Description
<p>Add Sample Comment</p>	<p>Opens the Add Sample Comment dialog box. You add a comment for the sample in the Add Comment field.</p> <p><i>Figure 8-19: Add Sample Comment dialog box</i></p>  <p>Note: If multiple comments have been added for a sample, then these comments are displayed 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.</p>
<p>Delete Sample Comment</p>	<p>Opens the Add Delete Comment dialog box. Click OK to delete the comment for the sample and close the dialog box.</p> <p><i>Figure 8-20: Delete Comment dialog box</i></p> 

Table 8-6: Sample tab, Sample Data sub-tab, Comments pane context menu (Continued)

Option	Description
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267 .
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 37 . 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 58 .
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 .
<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. If a patient is not associated with the sample, then Set Patient is available on the context menu for the pane and you can [associate](#) one. You can also 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

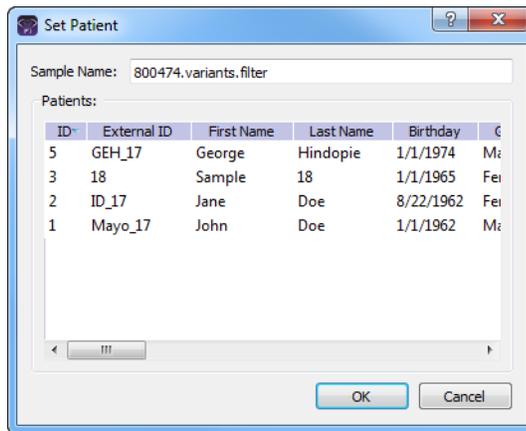
Option	Description
Add All To Custom Report	Selects all the information that is displayed for the selected patient for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267 .
Add Patient External ID to Custom Report	Selects only the External ID for the patient for inclusion in a custom report. See Chapter 10, “Geneticist Assistant Reports,” on page 267 .
Set Patient	Opens the Set Patient dialog box in which you can associate a patient with a sample. See “To associate a patient with a sample” below.

To associate a patient with a sample

1. If you have not already done so, do one of the following to open the Sample tab for the sample:
 - Right-click a sample entry on the Run tab, and on the context menu that opens, select Sample Details.
 - Double-click a sample entry on the Run tab.
2. Open the Sample Data sub-tab.
3. Right-click in the Patient pane, and on the context menu that opens, click Set Patient.

The Set Patient dialog box opens. The dialog box lists all the patients in your Geneticist Assistant database.

Figure 8-21: Set Patient dialog box



4. Select the patient, and then click OK.

The Set Patient dialog box closes. The patient information is displayed in the Patient pane.

Related Patients pane

The Related Patients pane displays all the relatives of the patient who is associated with the sample. Right-click a patient in the Related Patients pane to open a context menu with the following options:

Table 8-8: Sample 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 112. Note: You can also double-click a patient entry to open the Patient tab.

Table 8-8: Sample tab, Related Patients pane context menu (Continued)

Option	Description
Add New Relationships	The Add New Patient dialog box opens. By default, the Is Related to Existing Patient option is selected and unavailable. Add the needed relationships for the patient. See Step 3 of "To add new patients one at a time" on page 107.
Remove Patient's Relationships	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 267.
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see "To show/hide columns in a tab pane" on page 37.</p> <p>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 58.</p>
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.

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 126.



For detailed information about generating and saving both standard and custom Sample reports, see [Chapter 10, “Geneticist Assistant Reports,”](#) on page 267.

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 247.](#)
- [“Working with Reference Information Panes on the Variant Tab” on page 257.](#)

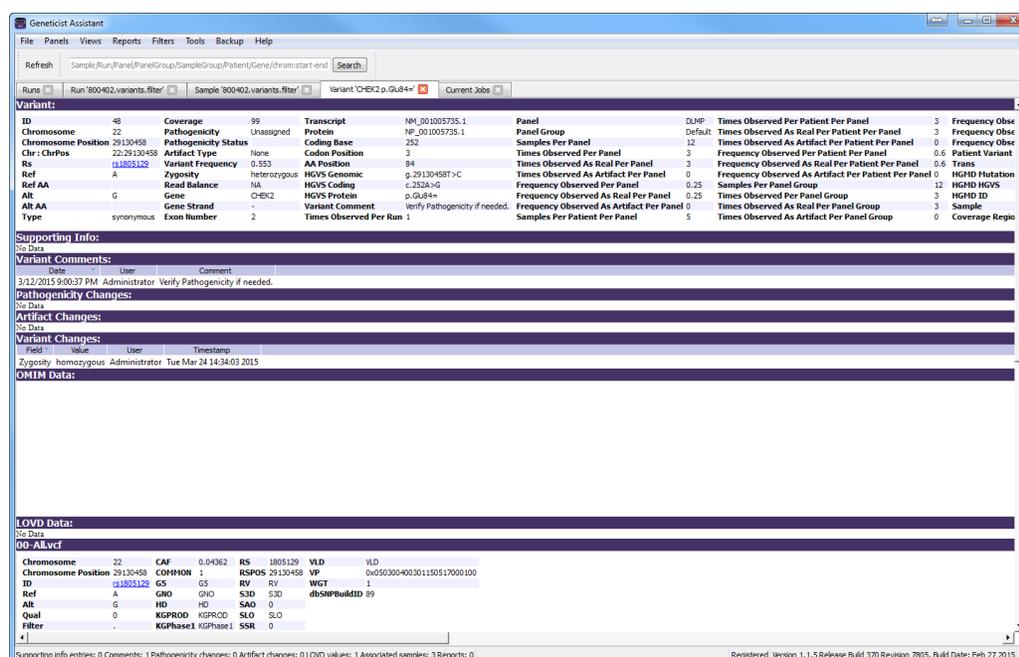
Chapter 9
Geneticist Assistant Data Review - Variant Tab

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



By default, the Variant tab displays 16 panes when it first opens. The panes are of two types: 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 248.](#)
- [“Reference information panes” on page 256.](#)



If any of the reference information panes are not displayed, contact SoftGenetics to download the latest reference files.



Because so many panes can be displayed on the Variant tab, remember, you can always select which panes to show and which to hide to make it easier for you to review variant data. See [“To show/hide a tab pane” on page 32](#).

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. By default, the following eight variant information panes are displayed on the Variant tab when it first opens:

- [“Variant pane” on page 249](#).
- [“Supporting Information pane” on page 251](#).
- [“Variant Comments pane” on page 252](#).
- [“Pathogenicity Changes pane” on page 253](#).
- [“Artifact Changes pane” on page 253](#).
- [“Variant Changes pane” on page 253](#).
- [“Associated Samples pane” on page 253](#).
- [“Variant’s Reports pane” on page 255](#).

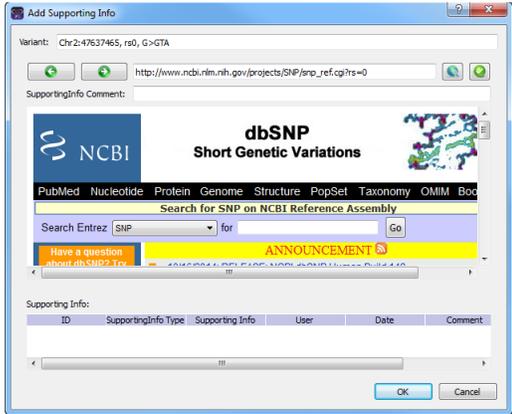
The following commands are displayed on the context menu for every variant information pane:

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 267 .
Load Columns	
Manually	<p>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.</p> <p>Note: For information about showing and hiding columns dynamically, see “To show/hide columns in a tab pane” on page 37.</p> <p>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 58.</p>
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.

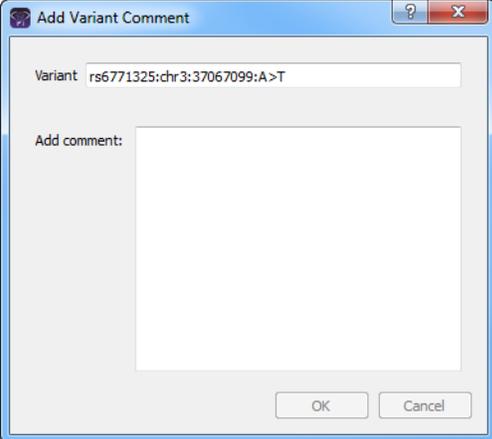
Option	Description
Load Filters	
Manually	Opens the Filters table for the pane. See “To filter the data that is displayed in a tab pane” on page 32.
<Filter Template Name> - Saved by <User Name>	Opens the saved Filter template for the pane. Limits the display in the pane to only the data (fields) that 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 218.](#)) You can right-click in the pane to open a context menu that, in addition to the command pane commands, has the following pane-specific commands:

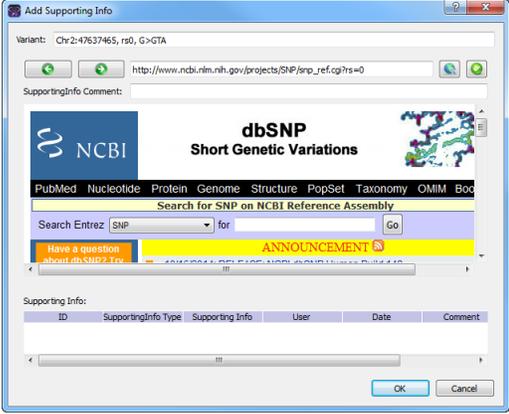
Option	Description
Submit Supporting Hyperlink	<p>Opens the Add Supporting Info dialog box, which, by default, displays the dbSNP web page for the variant. You can use the browser in this dialog box to select this web page as supporting information for the variant, or navigate to a different page.</p> <p><i>Figure 9-2: Add Supporting Info dialog box</i></p>  <p>Note: See Step 9 of “To add supporting information for a variant” on page 257.</p>

Chapter 9
 Geneticist Assistant Data Review - Variant Tab

Option	Description
Add Comment	<p>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.</p> <p><i>Figure 9-3: Add Variant Comment dialog box</i></p>  <p>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.</p>

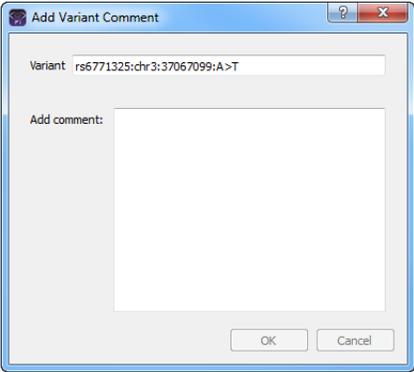
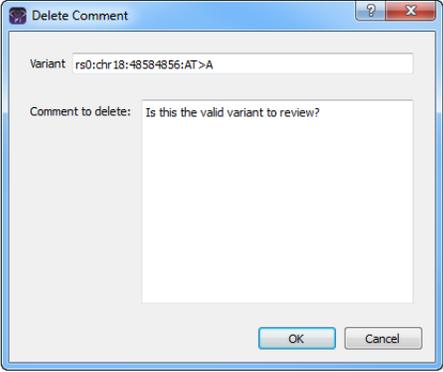
Supporting Information pane

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 257.](#)) You can right-click in the pane to open a context menu that, in addition to the command pane commands, has the following pane-specific commands:

Option	Description
<p>Submit Supporting Hyperlink</p>	<p>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.</p> <p><i>Figure 9-4: Add Supporting Info dialog box</i></p>  <p>Note: See Step 9 of “To add supporting information for a variant” on page 257.</p>

Variant Comments pane

The Variant Comments pane displays all the comments that have been added for the variant in reverse chronological order. You can right-click in the pane to open a context menu that, in addition to the command pane commands, has the following pane-specific commands:

Option	Description
<p>Add Comment</p>	<p>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.</p> <p><i>Figure 9-5: Add Variant Comment dialog box</i></p>  <p>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.</p>
<p>Delete Comment</p>	<p>Opens the Delete Comment dialog box. Click OK to delete the comment for the variant and close the dialog box.</p> <p><i>Figure 9-6: Delete Comment dialog box</i></p> 

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. For a description of the context menu options for the pane, see [“Variant information panes” on page 248](#).

Artifact Changes pane

The Artifact Changes pane displays the history of the changes that have been made to the artifact type for the variant in reverse chronological order. When a variant is first added to the Geneticist Assistant database, the Artifact Type is set to None. If the Artifact Type is set to Sequencing, for example, then this change and the name of the user who made the change, and the date and the time of the change are recorded and displayed in this pane. For a description of the context menu options for the pane, see [“Variant information panes” on page 248](#).

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. For a description of the context menu options for the pane, see [“Variant information panes” on page 248](#).

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 command pane commands, has the following pane-specific commands:

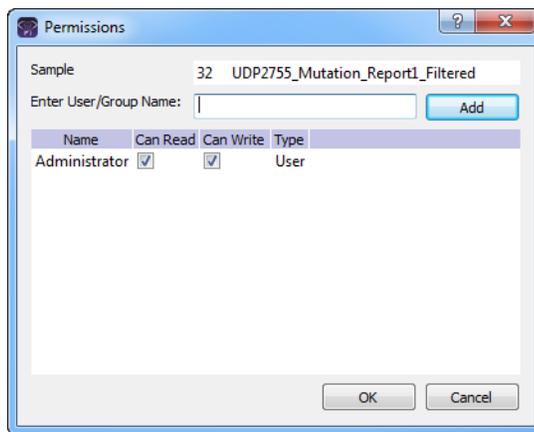
Option	Description
Sample Detail	Opens the Sample tab for the sample. See Chapter 8, “Geneticist Assistant Data Review - Sample Tab,” on page 215 .
Permissions	Opens the Permissions dialog box. You use the options on this dialog box to set or modify the permissions (Can Read or Can Read and Can Write) that the specified users or groups are to have for selected samples. If you are an Administrator user, then you can set or modify the permissions for a sample that any user has submitted. If you are a standard user, then you can set or modify the permissions for only those samples that you have submitted to the Geneticist Assistant database, or for those samples that other users have submitted and to which you have been granted Can Read and Can Write permissions. See “To set or modify the Read/Write permissions for the samples in which a selected variant was found” on page 254 .

To set or modify the Read/Write permissions for the samples in which a selected variant was found

1. If you have not already done so, open the Variant tab for the appropriate variant.
2. In the Associated Samples pane, select the sample, or CTRL-click to select multiple for which the permissions are being set.
3. Right-click any selected sample, and on the context menu that opens, click Permissions.

The Permissions dialog box opens.

Figure 9-7: Permissions dialog box (Variant tab, Associated Samples pane)



- The first time that the dialog box opens, if any sample was submitted within a run using a Geneticist Assistant version with a build date of 4/8/15 or later, then the names of the users who submitted the runs are displayed in the lower pane of the dialog box and both the Can Read and Can Write permissions are selected. If a standard user submitted any run, then the Administrator user name is also displayed.
 - For samples that were submitted within runs using earlier versions of Geneticist Assistant, the pane is blank. For subsequent openings, the dialog box displays the names of all the users and/or groups and their assigned permissions for the samples.
4. Modify the permissions for any user or group as needed and then click OK; otherwise, to add permissions for a new user or group, go to [Step 5](#).



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.

5. 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.



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.

6. Select the appropriate 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 Read permission is already assigned for the selected user or group. If just this permission is selected, then the user or group can simply view the sample records. They cannot modify them.

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

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 sample records.

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 126.



For detailed information about generating and saving both standard and custom Run reports, see [Chapter 10, “Geneticist Assistant Reports,”](#) on page 267.

Reference information panes

By default, the following eight reference information panes are always displayed when the Variant tab first 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 “To specify the OMIM settings” on page 65.
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 LOVD (Leiden Open Variation Database) world-wide 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).
Exome Aggregation Consortium (ExAC.r*.*.sites.vcf.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:

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 257.
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 260 or “To add a custom field to variant tables and panes” on page 262.

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 260.
- Add a custom field to variant tables and panes. See [“To add a custom field to variant tables and panes”](#) on page 262.

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 251.)

1. If you have not done so already, do one of the following to open the Variant tab:
 - Double-click a variant on the Variants sub-tab on the Patient tab.
 - Double-click a variant on the Variant sub-tab on the Panel Statistics tab.
 - Right-click a variant on the Sample Variant Data sub-tab on the Sample tab, and on the context menu that opens, select Variant Details.
 - Double-click a variant on the Sample Variant Data sub-tab on the Sample tab.
2. Do one of the following:
 - To reference information from any reference track, go to [Step 3](#).
 - To reference a specific web page, go to [Step 8](#).
3. In the appropriate reference information pane, review the information that is to be used for supporting information. See [“Reference information panes”](#) on page 256.



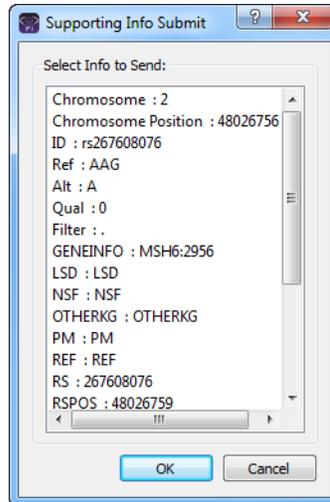
This information is included in the References folder. Use the scroll bar at the bottom of the pane to view the available reference information for the variant. Note that all reference information is not available for all variants.

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Geneticist Assistant Data Review - Variant Tab

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 9-8: Supporting Info Submit dialog box

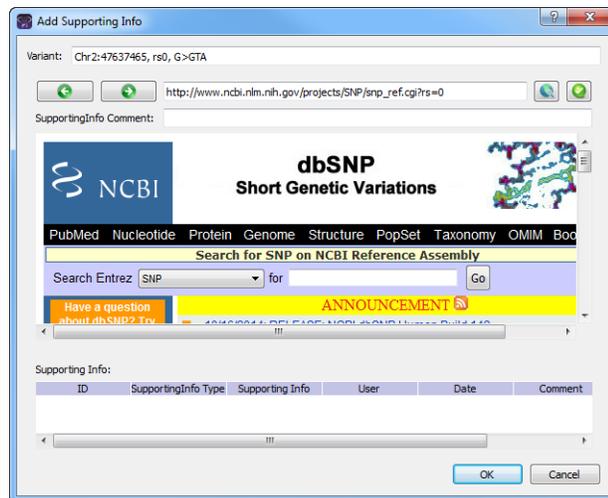


5. Select the information that is to be used as supporting information, and then click OK.
The Add Supporting Info dialog box closes. The information is displayed in the Supporting Information pane. See [“Supporting Information pane” on page 251](#).
6. Repeat [Step 3](#) through [Step 5](#) for each piece of reference information that is to be added as supporting information.
7. Optionally, continue to [Step 8](#) to also add supporting information for the variant from a web page.

- Right-click anywhere on the Variant pane or the Supporting Information pane, and on the context menu that opens, click Submit Supporting Hyperlink.

The Add Supporting Info (for a web page) dialog box opens. By default, the dialog box displays the dbSNP page for the variant.

Figure 9-9: Add Supporting Info dialog box



- 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.

- 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 251.

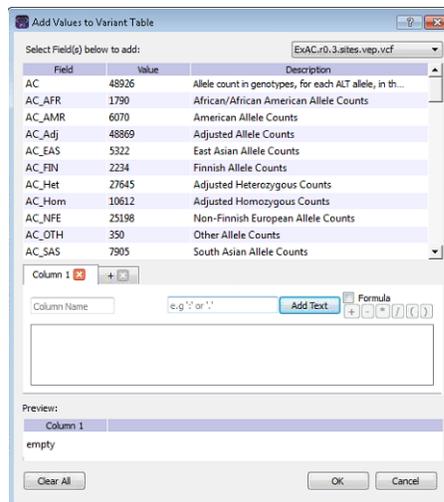
To add fields from a reference database to variant tables and panes

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.

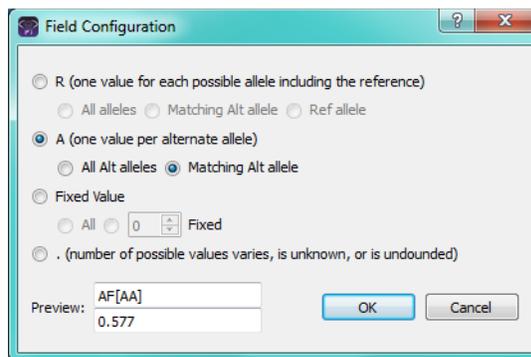
Figure 9-10: Add Values to Variant Table dialog box



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 9-11: Field Configuration dialog box



4. Select the values in the field that are to be added to the various panes and tables.

Value	Description and Options
R (one value for each possible allele including the reference)	<p>The field contains multiple values, with one value for each possible allele, including the reference allele.</p> <ul style="list-style-type: none"> • 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)	<p>The field can contain multiple values, with one value for each alternate allele.</p> <ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • All - Add all values (multiple values) • Fixed - Add only the specified number of fixed values. If 1 is specified, then this is a single value; otherwise, 2 or greater adds multiple values.
. (number of possible values varies, is unknown, or is unfounded)	<p>Based on different variants, the number of values in the field can vary.</p>

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

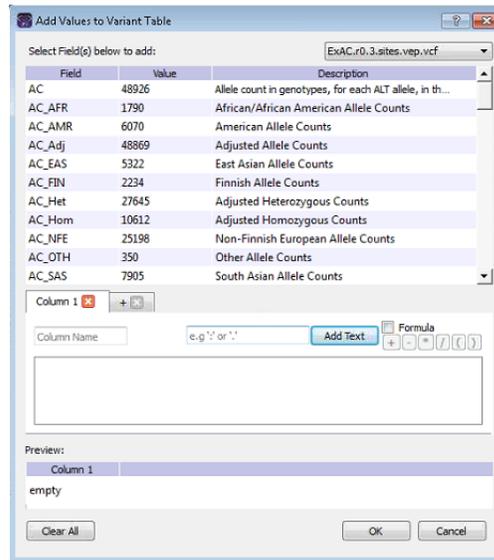
You can combine multiple fields in a reference database to create a custom field, and then add the custom field and its value to the Variant pane and the Variants pane on the Sample Variant Data sub-tab on the Sample tab. When you select a reference database field to use for creating a custom field, 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.

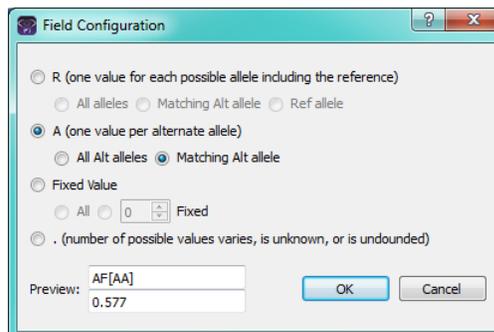
Figure 9-12: Add Values to Variant Table dialog box



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 is to be used for the custom formula.

Figure 9-13: Field Configuration dialog box



4. Select the value in the field that is to be used in the formula for the custom field.

Value	Description and Options
R (one value for each possible allele including the reference)	<p>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:</p> <ul style="list-style-type: none"> • 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)	<p>The field can contain multiple values, with one value for each alternate allele. Therefore, the values that you can select are the following:</p> <ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • Fixed - Add only the specified number of fixed values. If 1 is specified, then this is a single value

5. Click OK.



After you select the value that is 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 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.

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Geneticist Assistant Data Review - Variant Tab

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. If you want to generate an output file of the data, then you have two options. You can generate a standard report, or you can select some or all of the data for inclusion in a custom report.

This chapter covers the following topics:

- [“Generating a Standard Report” on page 269.](#)
- [“Generating a Custom Report” on page 273.](#)

Chapter 10
Geneticist Assistant Reports

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 179.
 - [“Coverage Regions tab”](#) on page 192.
 - The Panel Statistics tab: See [“Reviewing Panel Statistics”](#) on page 98, [“To calculate panel statistics \(Runs tab\)”](#) on page 163 or [“To calculate panel statistics \(Run tab\)”](#) on page 197.
 - [Chapter 8, “Geneticist Assistant Data Review - Sample Tab,”](#) on page 215.
 - [Chapter 9, “Geneticist Assistant Data Review - Variant Tab,”](#) on page 245.



To generate an Action List report, you must first create the Action List to open the Action List tab. See [“To add selected panel regions or variants to an Action List” on page 195](#) or [“To add selected panel regions or variants to an Action list” on page 235](#).

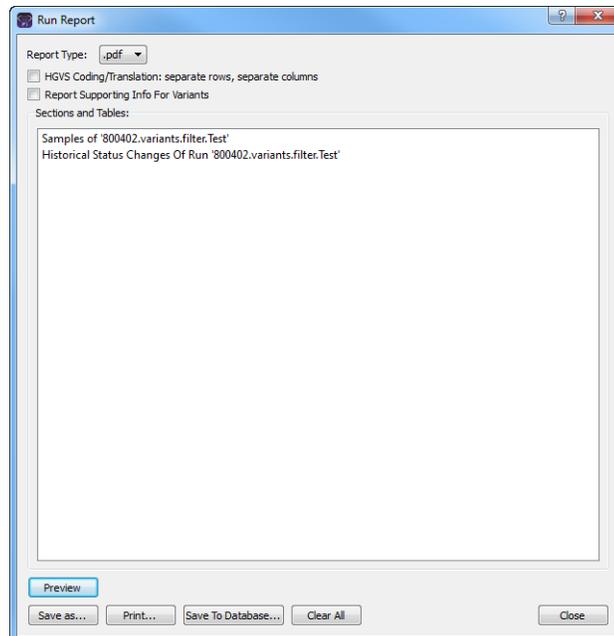
2. Do one of the following:

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



- 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 37.

- Select the report type.



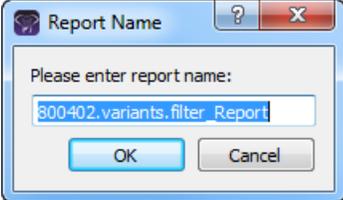
Four formats are available—PDF (the default value), HTML, XML, or CSV, which is a comma-separated values file.

- 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.
- Do any or all of the following as needed:

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: <ul style="list-style-type: none"> 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
<p>Save to Database</p>	<p>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.</p> <p><i>Figure 10-2: Report Name dialog box</i></p>  <p>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.</p>
<p>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.</p>	

7. Click Close to close the Report dialog box.

Generating a Custom Report

If you want to generate a custom output file of the data after a run is successfully completed, you can [generate](#) a *custom* report. A custom report contains only selected data that you specify from a data tab or tabs.

To generate a custom report

1. Open the appropriate tab. See:
 - [Chapter 7, “Geneticist Assistant Data Review - Run Tab,”](#) on page 179.
 - [“Coverage Regions tab”](#) on page 192.
 - The Panel Statistics tab: See [“Reviewing Panel Statistics”](#) on page 98, [“To calculate panel statistics \(Runs tab\)”](#) on page 163 or [“To calculate panel statistics \(Run tab\)”](#) on page 197.
 - [Chapter 8, “Geneticist Assistant Data Review - Sample Tab,”](#) on page 215.
 - [Chapter 9, “Geneticist Assistant Data Review - Variant Tab,”](#) on page 245.



To generate an Action List report, you must first create the Action List to open the Action List tab. See [“To add selected panel regions or variants to an Action List”](#) on page 195 or [“To add selected panel regions or variants to an Action list”](#) on page 235.

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.

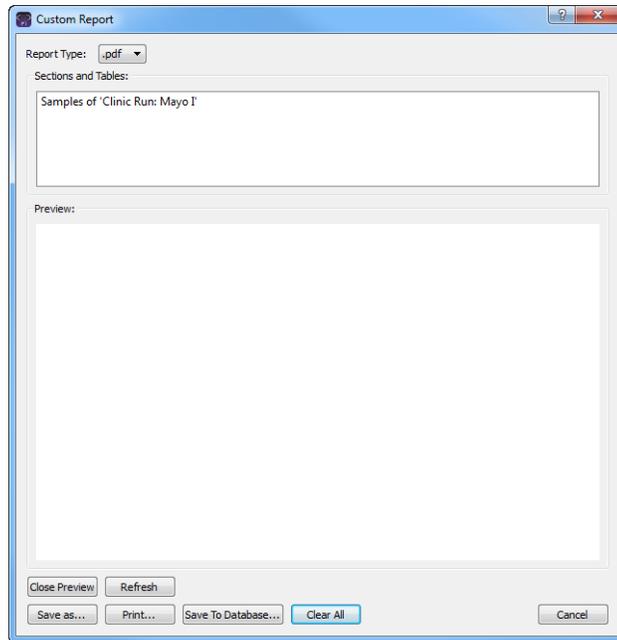


For both options, only the columns that are shown in the pane can be selected for inclusion in the report. Hidden data columns cannot be selected. See [“To show/hide columns in a tab pane”](#) on page 37.

3. On the Geneticist Assistant main menu, click Reports > Custom Report.

The Custom Report dialog box opens. All the data that is to be included in the report is displayed in the Sections and Tables pane of the dialog box. See [Figure 10-3 on page 274](#).

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.

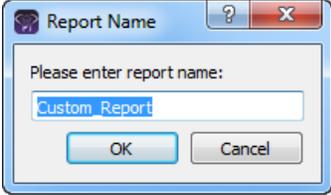
5. Select the report type.



Four formats are available—PDF (the default value), HTML, XML, or CSV, which is a comma-separated values file.

6. Do any or all of the following as needed:

Table 10-3: Custom report options

Option	Description
Click Preview	<p>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:</p> <ul style="list-style-type: none"> • Close Preview—Close the Preview and leave a blank Report dialog box open. • Refresh—Refresh the preview in real time.
Click Save As	<p>To save the report. You must specify the name of the report and the location for the saved report.</p>
Click Print	<p>To print the report.</p>
Save to Database	<p>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.</p> <p><i>Figure 10-4: Report Name dialog box</i></p>  <ol style="list-style-type: none"> 1. Rename the report. 2. Click OK. <p>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.</p>

7. Click Clear All.



This ensures that information that is selected for a new custom report is not simply appended to an existing report.

8. Click Close to close the Report dialog box.

Chapter 10
Geneticist Assistant Reports

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 managing pathogenicity changes, a tool for specifying pathogenicities for specific variants in a panel, and a tool for combining BAM files into a single sample.

This chapter covers the following topics:

- [“Building a BED File with the BED File Builder Tool” on page 279.](#)
- [“Deleting Pathogenicity Changes with the Delete Pathogenicity Changes Tool” on page 283.](#)
- [“Importing Variants with the Import Variants Tool” on page 285.](#)
- [“Merging BAM Files with the Merge BAM Files Tool” on page 288.](#)

Chapter 11
Geneticist Assistant Tools

Building a BED File with the BED File Builder Tool

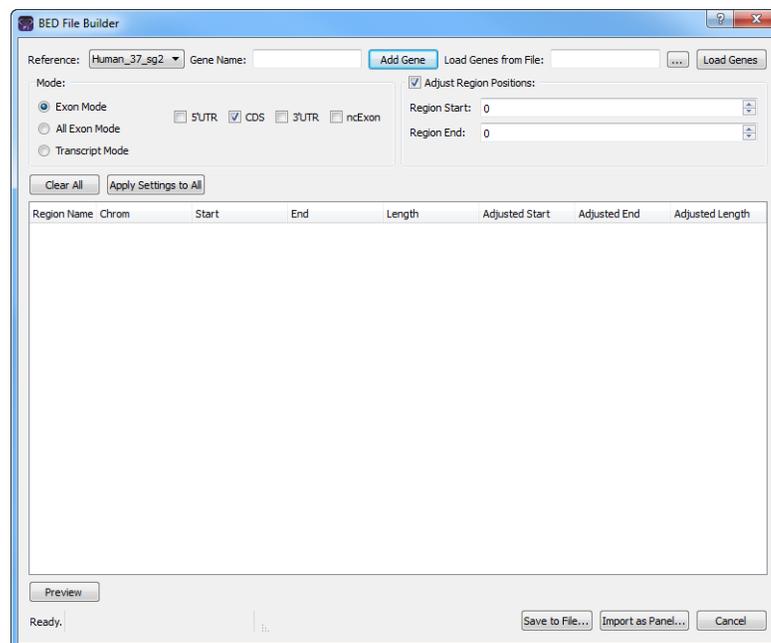
You use BED files to create panels for Geneticist Assistant. A BED file defines the genomic regions that are targeted for your particular study or test, which are referred to as *panel regions*. If do not have a BED file for a panel, you can use the BED File Builder tool to [build](#) one based on the information that you specify.

To build a BED file with the BED File Builder tool

1. On the Geneticist Assistant main menu, click Tools > BED File Builder.

The BED File Builder dialog box opens.

Figure 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 **ASHIL-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.

- 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 <i>not</i> 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 <i>not</i> included in Exon mode.

- Do one of the following:

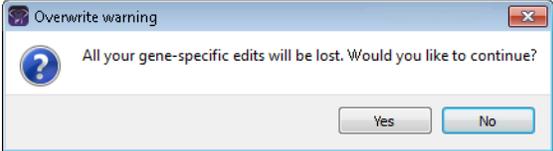
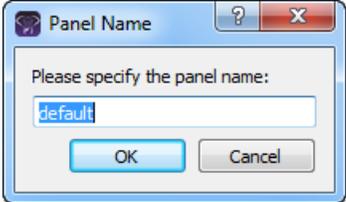
- For Exon mode and All Exon mode—Select the appropriate transcript, or click the Expand icon  next to the transcript, and then select the coding region and/or untranslated regions for building the BED file.

Figure 11-2: BED File Builder tool, Exon mode

Region Name	Chrom	Start	End	Length
BRCA1	17	41196312	41277500	81189
NM_007294.3	17	41196312	41277500	81189
5'UTR	17	41196312	41197694	1383
Coding Regions	17	41197695	41267796	70102
3'UTR	17	41277288	41277500	213
NM_007297.3	17	41196312	41277408	81157
NM_007308.2	17	41106312	41276122	70821

- For Transcript mode—Select the appropriate transcript.

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

Option	Steps
<p>To open the Preview pane and view a preview of the BED file</p>	<p>Click Preview. The information for each region (Chromosome, Region Start, Region End, Region Name, Score, and Strand) is displayed in the preview.</p> <p>Note: For detailed information about the fields that are displayed in the preview, see https://genome.ucsc.edu/FAQ/FAQformat.html#format1.</p> <p>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.</p>
<p>To adjust the region start and end positions</p>	<p>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</p>
<p>To apply the current settings (the selected mode and the Start/End Region values) to all genes, including any previously added genes</p>	<ol style="list-style-type: none"> 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. <p><i>Figure 11-3: Overwrite warning message</i></p>  <ol style="list-style-type: none"> Click Yes to close the message and overwrite all the current gene-specific settings with the current gene settings.
<p>To immediately use the BED file as a panel</p>	<ol style="list-style-type: none"> Click Import as Panel. The Panel Name dialog box opens. <p><i>Figure 11-4: Panel Name dialog box</i></p>  <ol style="list-style-type: none"> 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. Continue to “Importing a Panel” on page 85.

Chapter 11
Geneticist Assistant Tools

6. Click OK.

The Save As dialog box opens. By default, the BED file is named default.bed.

7. Modify the name of the BED file as needed, and save it in a location of your choosing.

Deleting Pathogenicity Changes with the Delete Pathogenicity Changes Tool

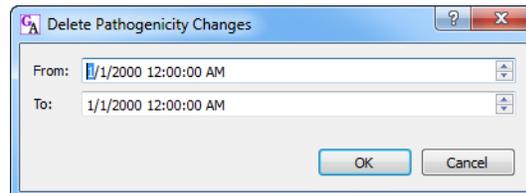
Sometimes, you might determine that a pathogenicity change or changes might not be valid. You use the Delete Pathogenicity Changes Tool to **delete** all the historical pathogenicity changes for all variants *that were made during a certain time period*. After you delete a historical pathogenicity change 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 11-5: Delete Pathogenicity Changes dialog box



2. Change the From value, the To value, or both values as needed. To change the value, select the value that is to be changed, for example, the month, and then you can manually enter the new value, or you can use the Up/Down field arrows to change the value.

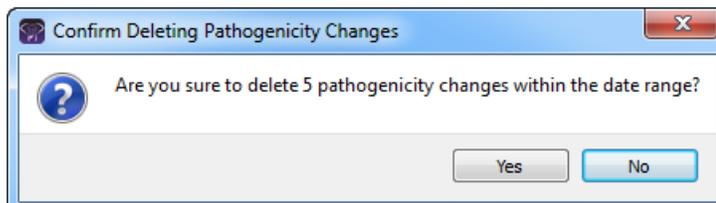


You must select each value that is to be changed one at a time. If you need to confirm the time period, remember that the Pathogenicity Changes pane on the Variant tab displays the date and time that a pathogenicity change was made. See “Pathogenicity Changes pane” on page 253.

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 11-6: 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

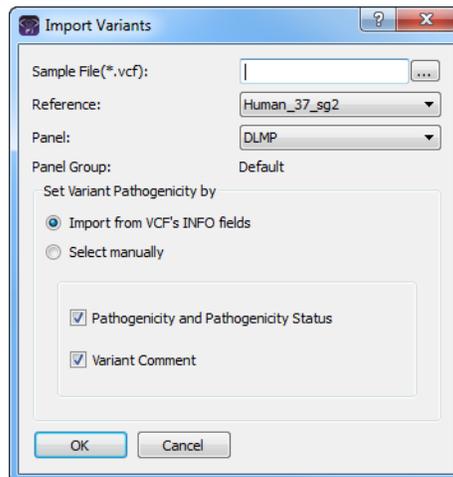
You can use the Import Variant Tool to [import](#) all the variants that are contained in a VCF file into your Geneticist Assistant database. If the VCF file contains the pathogenicity and pathogenicity status for the variants and/or comments for the variants, then you can also import this information. If the VCF file's INFO fields do not contain this information, then you can still use the tool to import the variants that are contained in the file, but then you can manually set the pathogenicity and/or pathogenicity status for the variants, as well as add a comment for the variants. For either situation, this means that if multiple variants have the same pathogenicity for a panel group, you do not have to set the pathogenicity of each variant individually. Instead, you can set the same pathogenicity for all the variants at once using this tool.

To import variants with the Import Variants tool

1. On the Geneticist Assistant main menu, click Tools > Import Variants.

The Import Variants dialog box opens.

Figure 11-7: Import Variants dialog box



2. Next to the Sample File field, click the Browse button  to open the Select a Variant File dialog box, and then browse to and select the appropriate VCF file.
3. On the Reference dropdown list, select the appropriate reference.
4. On the Panel dropdown list, select the panel for which you are settings the variants' pathogenicity.

The Panel group name is displayed for Panel Group.

5. Continue to one of the following:
 - [“To automatically set the variants’ pathogenicities and statuses from the VCF file”](#) below.
 - [“To manually set the variants’ pathogenicity and status”](#) below.

To automatically set the variants’ pathogenicities and statuses from the VCF file

1. Leave Import from VCF’s INFO fields selected.
2. Do either or both of the following as needed:
 - If the VCF file contains the pathogenicity and pathogenicity status for each variant, then to import this information, leave the Pathogenicity and Pathogenicity Status option selected. Otherwise, if the VCF file does not contain this information, or you do not want to import this information, then clear this option.
 - If the VCF file contains comments for the variants, then to import this information, leave the Variant Comment option selected. Otherwise, if the VCF file does not contain this information, or you do not want to import this information, then clear this option.
3. Click OK.

The Import Variants dialog box closes. A Current Jobs tab opens that shows the progress of importing the VCF file. The tab remains open after the import is complete. The variants with the selected information are now imported into the Geneticist Assistant database. These variants will now be displayed with the imported information (such as the pathogenicity) in any samples that contain these variants.

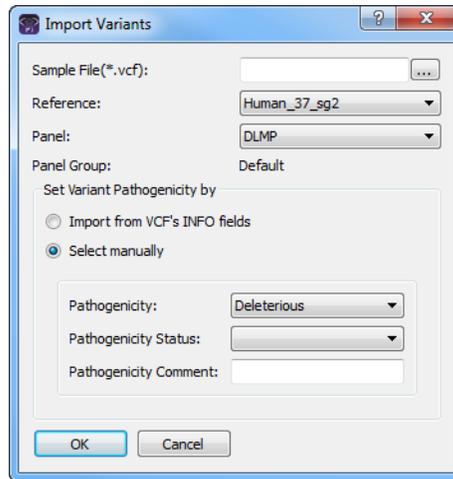
To manually set the variants’ pathogenicity and status

When you manually set the pathogenicity and status for the variants in the VCF file, then the pathogenicity and status are set to the *same* values for all the variants in the file. If you add a comment, then the same comment is added for all the variants.

1. Select the Select Manually option.

The Import Variants dialog box is updated with options for setting the pathogenicity and the pathogenicity status for the variants as well entering a common comment for the variants. See [Figure 11-8 on page 287](#).

Figure 11-8: Import Variants dialog box, Select manually option



2. Do any or all of the following as needed:
 - On the Pathogenicity dropdown list, select the pathogenicity for the variants.
 - On the Pathogenicity Status dropdown list, select the pathogenicity status for the variants.



Remember, you have the option of specifying custom pathogenicity types. Also, by default, only two statuses are available—Pending or Confirmed. You have the option of adding custom pathogenicity statuses. See “Managing Objects” on page 49.

- In the Pathogenicity Comment field, enter a common comment for all the variants.
3. Click OK.

The Import Variants dialog box closes. A Current Jobs tab opens that shows the progress of importing the VCF file. The tab remains open after the import is complete. The variants with the selected information are now imported into the Geneticist Assistant database. These variants will now be displayed with the specified information (such as the pathogenicity) in any samples that contain these variants.

Merging BAM Files with the Merge BAM Files Tool

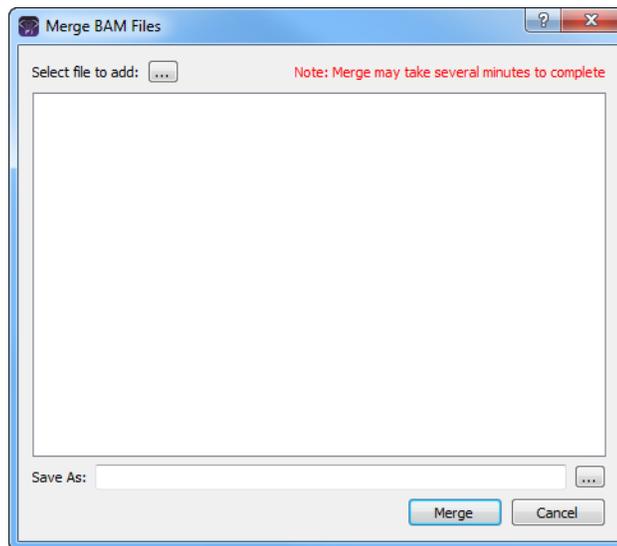
If you have multiple BAM files for a sample, you do not need to submit each file as an individual sample, and then assign each file to the same patient. Instead, you can use the Merge BAM Files tool to [merge](#) all the BAM files into one file and then submit the file as a single sample.

To merge BAM files

1. On the Geneticist Assistant main menu, click Tools > Merge BAM Files.

The Merge BAM Files dialog box opens.

Figure 11-9: 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 11
Geneticist Assistant Tools

Glossary

A

Action List

A list of items that require some type of follow up. In Geneticist Assistant, an Action List can be created for coverage regions on the Coverage 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 the application. At a minimum, the file contains the following information:

- Field #1 - Chromosome number for the region
- Field #2 - Chromosome start position
- Field #3 - Chromosome end position
- Field #4 - Optional comment column

C

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.

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.

D

dbNSFP pane

Found on the Variant tab. Displays the information from the dbNSFP database for a variant. See <https://sites.google.com/site/jpopgen/dbNSFP>.

dbSNP pane

Found on the Variant tab. Displays the information from the dbSNP database for a variant. See http://www.ncbi.nlm.nih.gov/SNP/get_html.cgi?whichHtml=overview.

E**Exome 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.

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.

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.

Missed Clinical Variant

A clinically relevant position that was identified in the Clinical Variants (ClinVar) database from the NCBI, and that was not covered for the sample.

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.

Reference Directory

Contains the information for the reference that was used for aligning data and/or detecting variants.

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.

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.

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 the application in a read-only capacity. Three types of users are available in Geneticist Assistant—an Administrator user, a Staff user, and a standard user.

V**Variant Frequency**

The frequency at which a variant is detected in a sample. In Geneticist Assistant, any value ≤ 1 is allowed, with 1 = 100%, 0.5 = 50%, and so on.

Variant Preferences

Determines how information about the variants is displayed in all the variant sub-tabs or panes in Geneticist Assistant. Set on the Variant Preferences tab for the Settings dialog box.

Z

Zygoty

Indicates whether the variant is heterozygous or homozygous. In Geneticist Assistant, the selected zygoty is based on the Zygoty settings that are specified on the Variant Preferences tab of the Settings dialog box.

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