

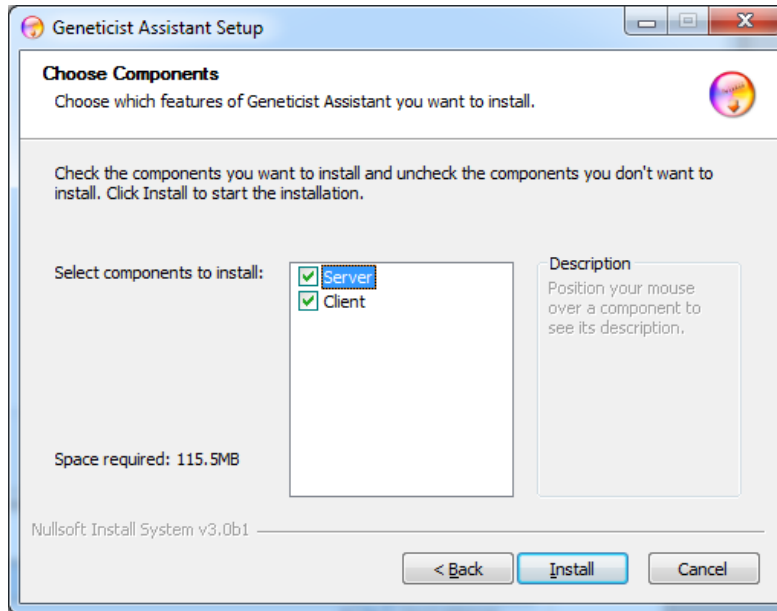
Geneticist Assistant Quick Start Guide

• Download

- Download GeneticistAssistantSetup_*.exe from the SoftGenetics ftp server.
- Contact tech_support@softgenetics.com for download information if you don't have it.

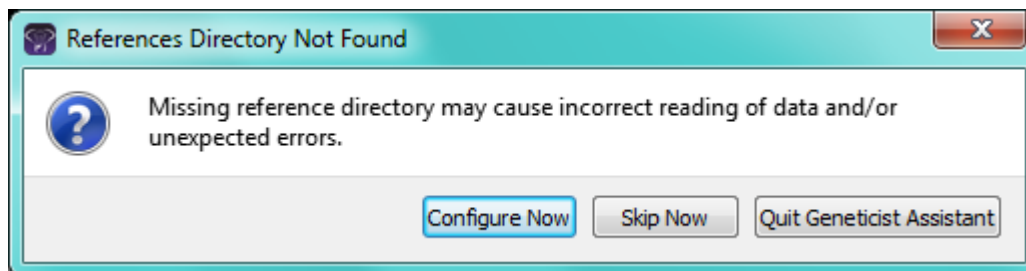
• Install

- Run GeneticistAssistantSetup_*.exe
- Install the server and client on the computer you wish to run the server from.
- Install just the client on other computers



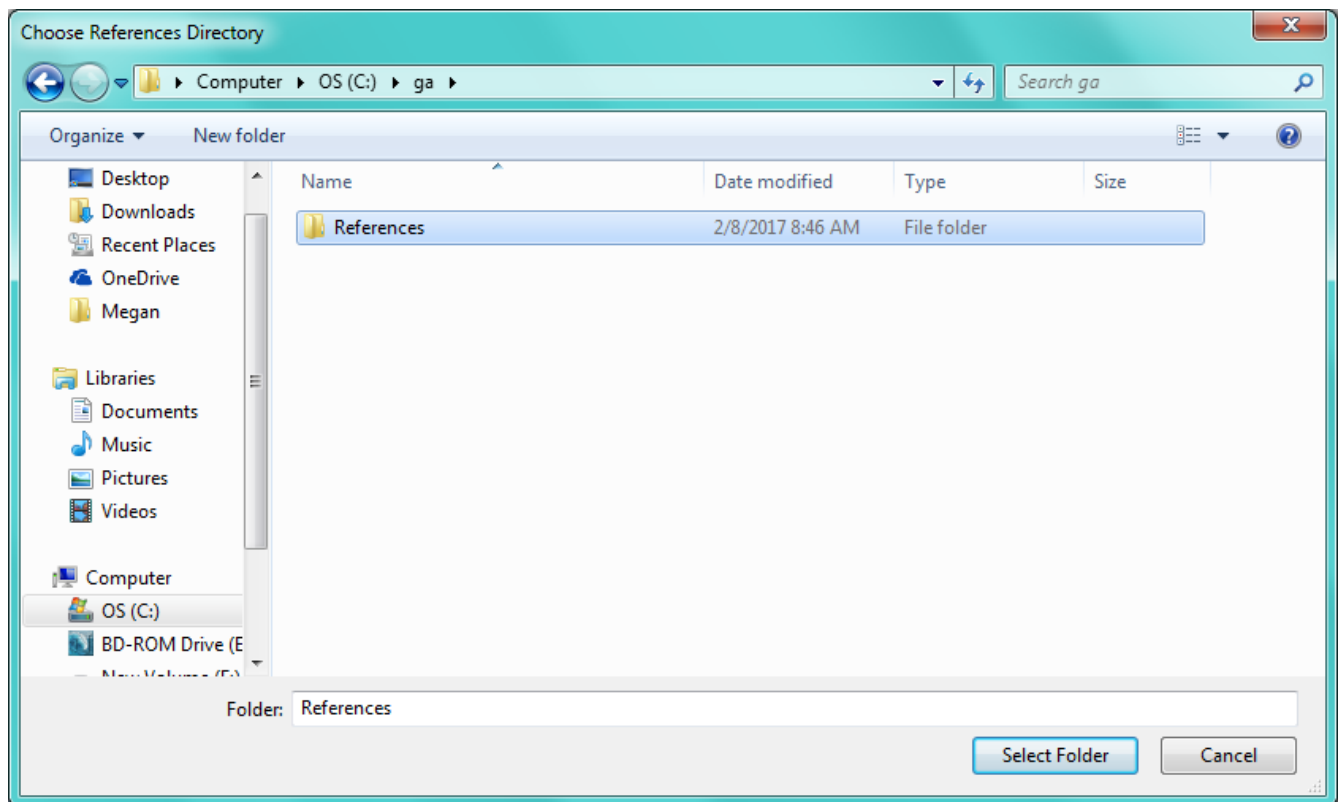
• Configure Reference Directory

- After opening Geneticist Assistant, you will be prompted to configure the reference directory - Click "Configure Now"

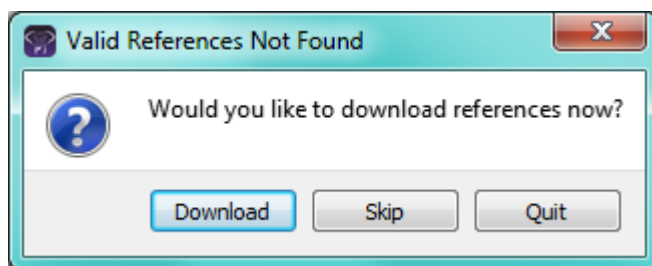


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- Browse and create a directory to store the reference
 - ✦ C:\ga\references

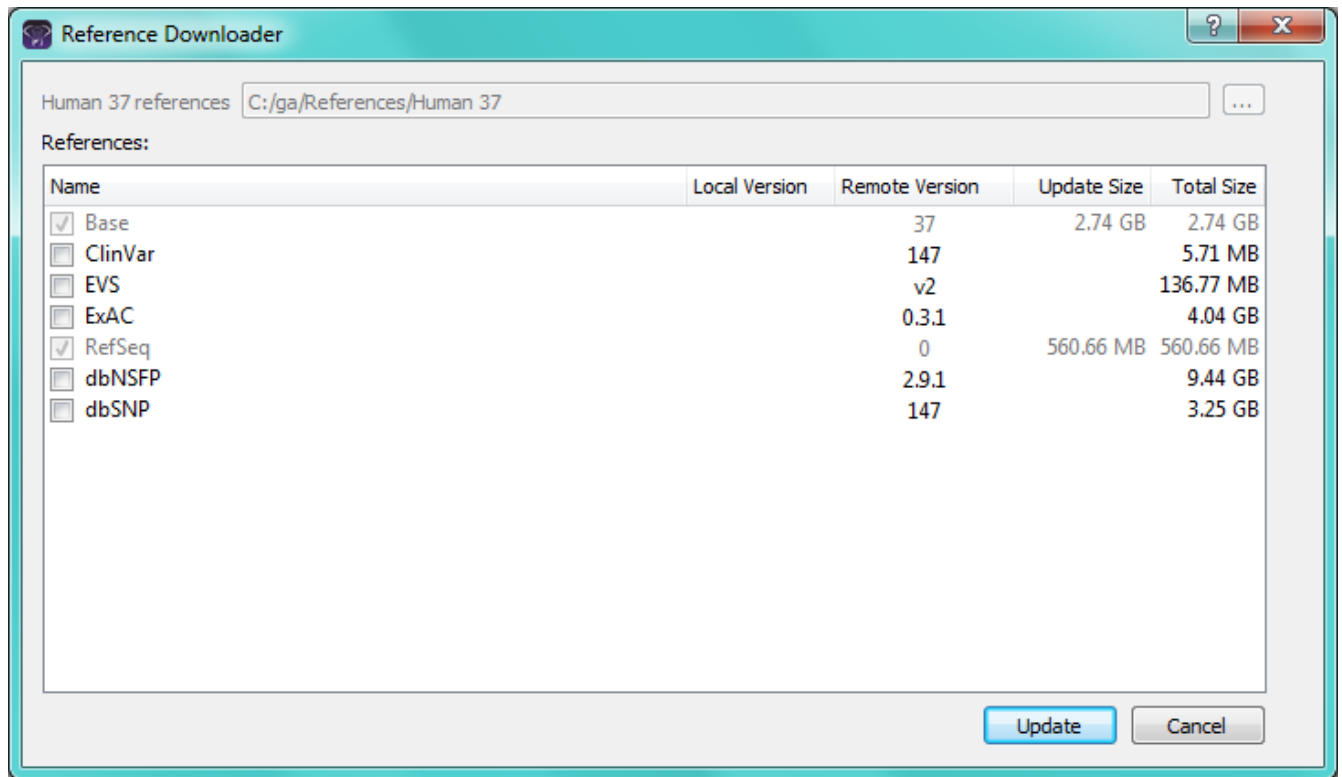


- Select to download reference



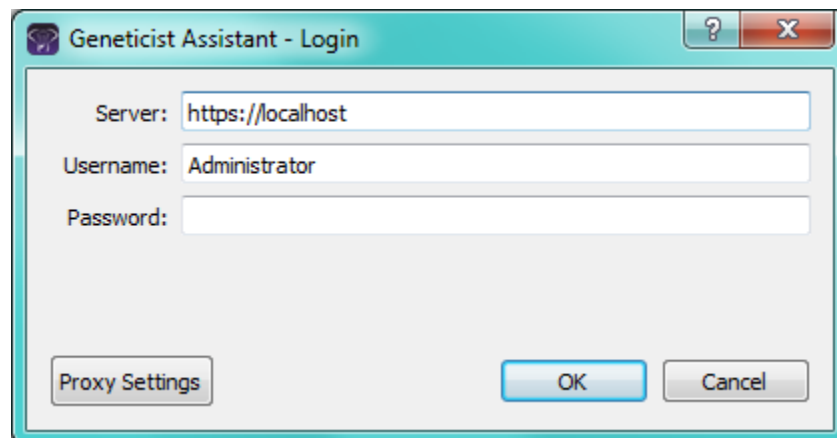
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- ↘ Select variant annotation databases for download



- ↘ Login

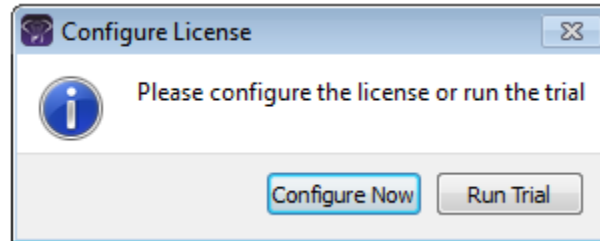
- ✦ Server: Name of the computer running the server
- ✦ Username: The user name you set up during install
- ✦ Password: The password you set up during install



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• Configure License

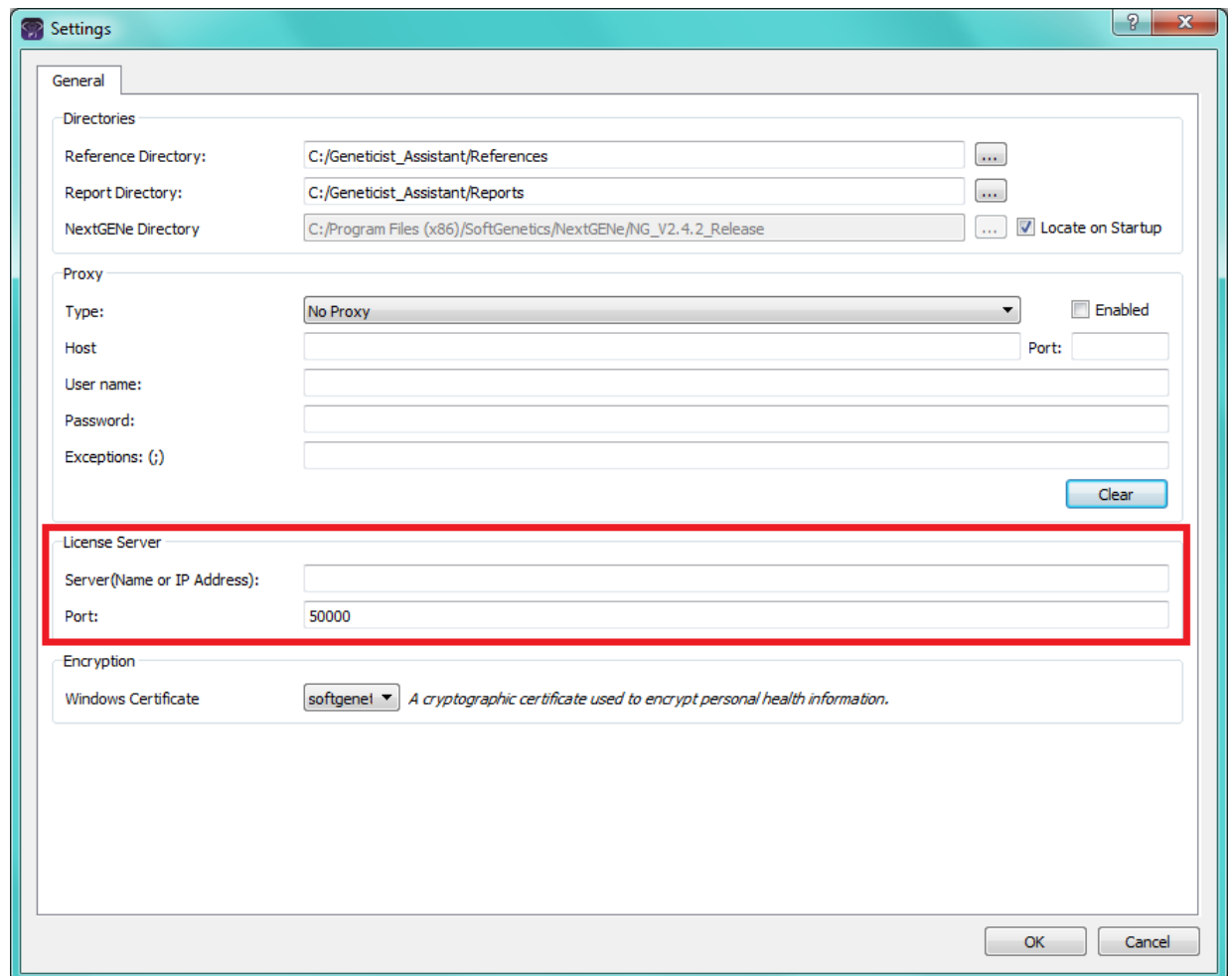
↳ Registered user click "Configure Now". All others click "Run Trial"



↳ Set the server to the computer with the License Server installed

✦ The default port is 50000

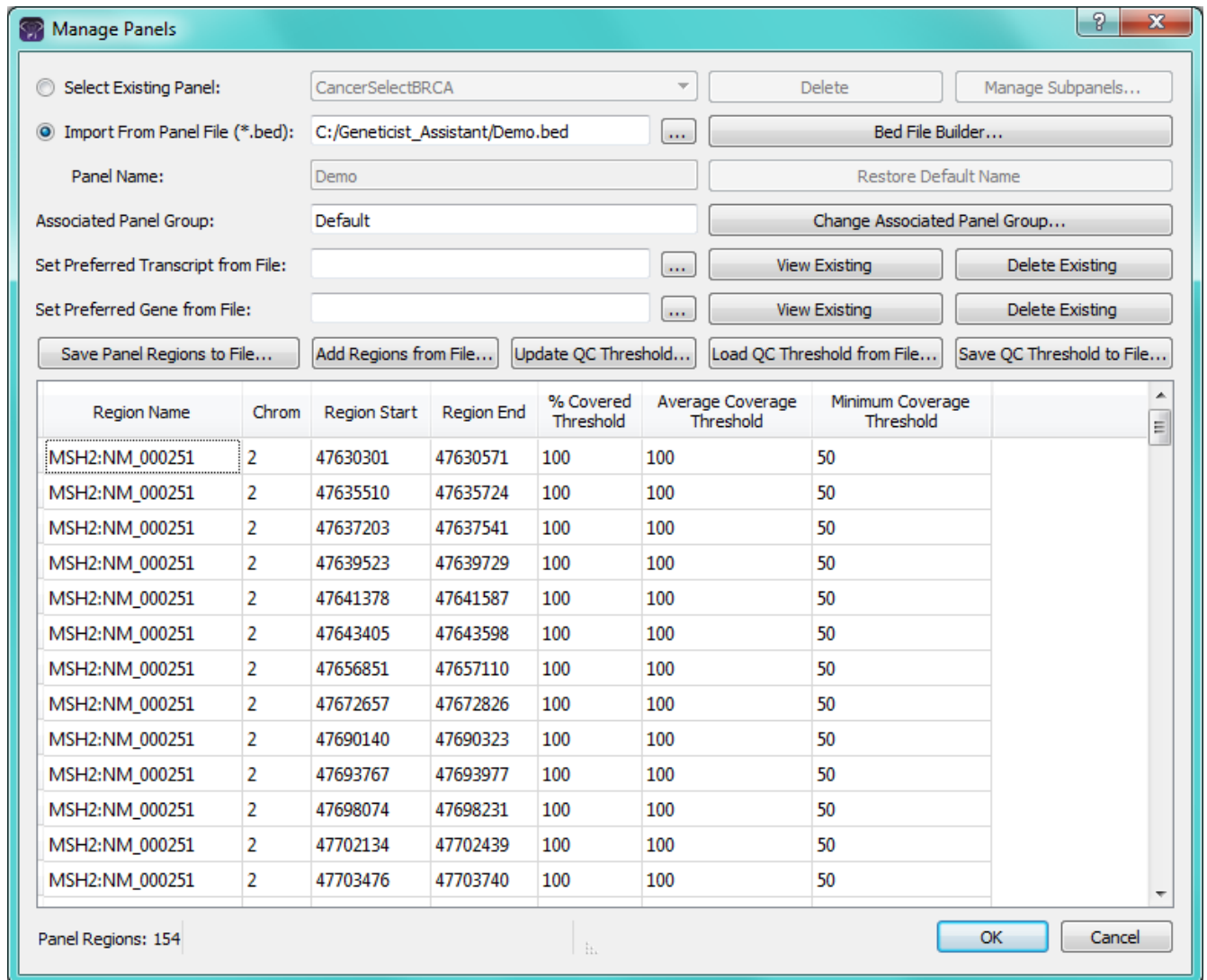
✦ Another guide that details how to install the License Server is available



Geneticist Assistant Quick Start Guide

• Create Panel

- Go to: Panels->Manage Panels
- Select "Import From Panel File (*.bed)"
- Click "...” to browse to and select a BED file to add as panel
- Click "Import Panel" to load BED file regions
- Click “OK” to import the panel



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• Submit New Run

- File > New Run
- Enter a name for the run
- Click "Select Variant and/or Coverage Files" to browse to and select your data files.
 - ✦ You can select multiple BAM and VCF files at the same time. They will be automatically paired based on their filename.
- Click "OK" to submit the samples.

New Run

Run Name: Demo

Required Settings:

Chemistry: Default

Instrument: Default

Reference: Human 37

Panel: Demo

Panel Group: Default

Case: None

Owner: Administrator

Multiple Samples in VCF

Optional Settings:

Run Date/Time: 2/8/2017 9:10 AM

User Group:

Sample Group:

Subpanel:

Identify Patient ID from Sample by Separator
_(underscore)

Import Pathogenicity from VCF

Submit Hotspots

Accepted VCF filters: PASS

:800418.igv-sorted_Output_Mutation_Report1_filtered:

Sample Name: 800418.igv-sorted_Output_Mutation_Report1_filtered

Variant File (*.vcf): _Assistant/Demo_data/800418.igv-sorted_Output_Mutation_Report1_filtered.vcf

Coverage/Pile Up File (*.bam): C:/Geneticist_Assistant/Demo_data/800418.igv-sorted_Output.bam

Reference: Human 37

Panel: xgen-pan-cancer-probes Panel Group: Default

Case: None

Run Date/Time: 2/8/2017 9:11 AM

Sample Group:

Subpanel:

Patients:

Import Pathogenicity from VCF

Submit Hotspots

Accepted Filters: PASS