

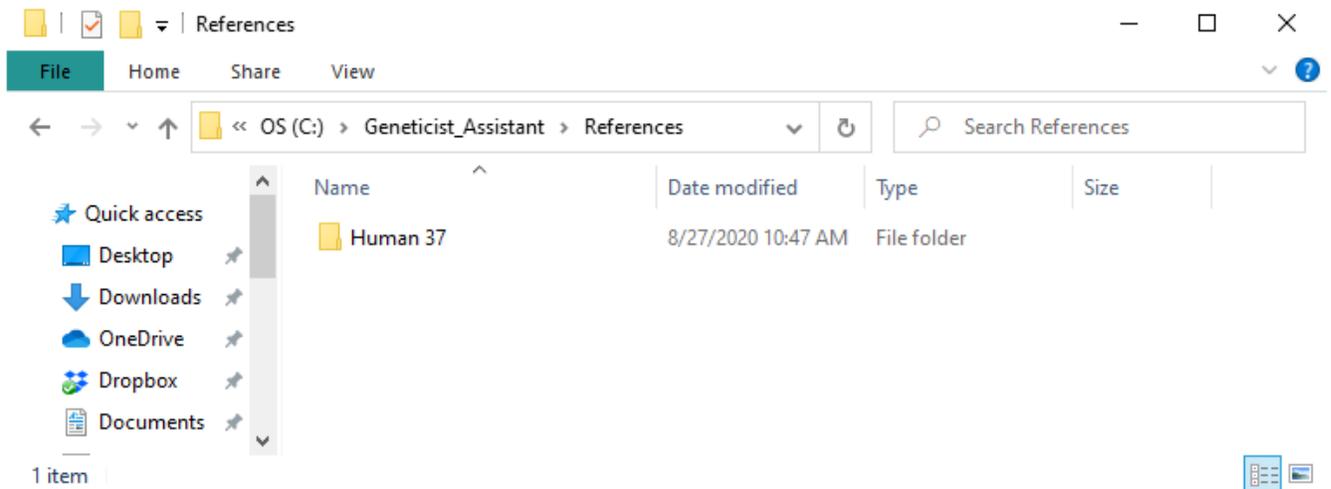
Geneticist Assistant Quick Start Guide

• Download

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

• Create Reference folder

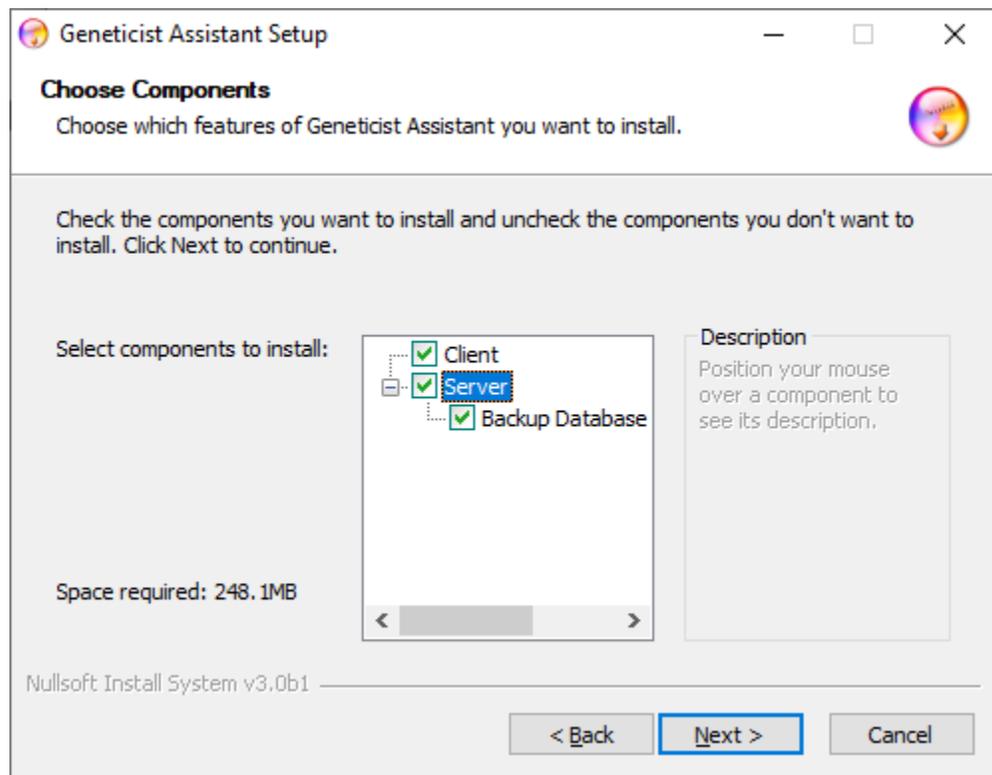
- Create a folder anywhere on your computer for the Geneticist Assistant reference files.
- Extract the downloaded references_minimal.zip file and copy the resulting Human 37 folder into the reference folder created.



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• Install

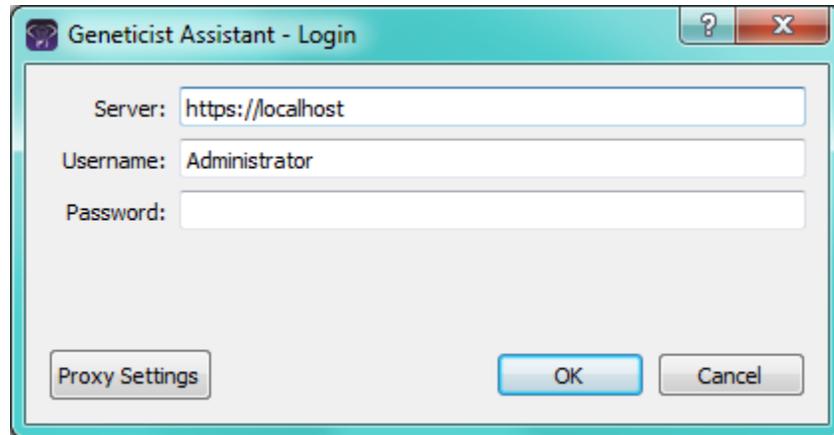
- Run GeneticistAssistantSetup_*.exe
- Install the server and client on the computer you wish to run the server from.
- Install just the client on other computers



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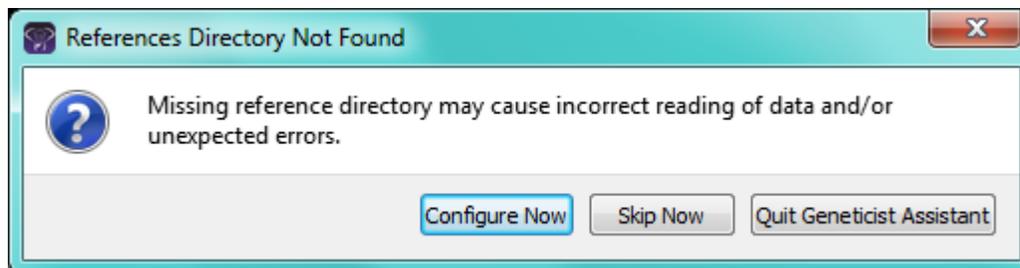
• Log in

- 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



• Configure Reference Directory

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

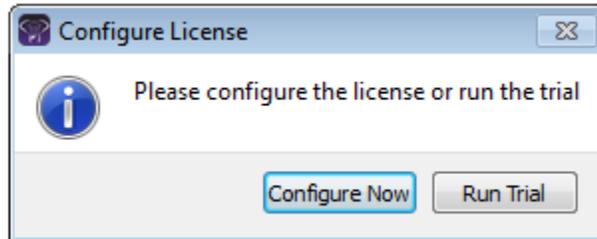


- Browse to and select the directory you created containing the Human 37 reference folder.

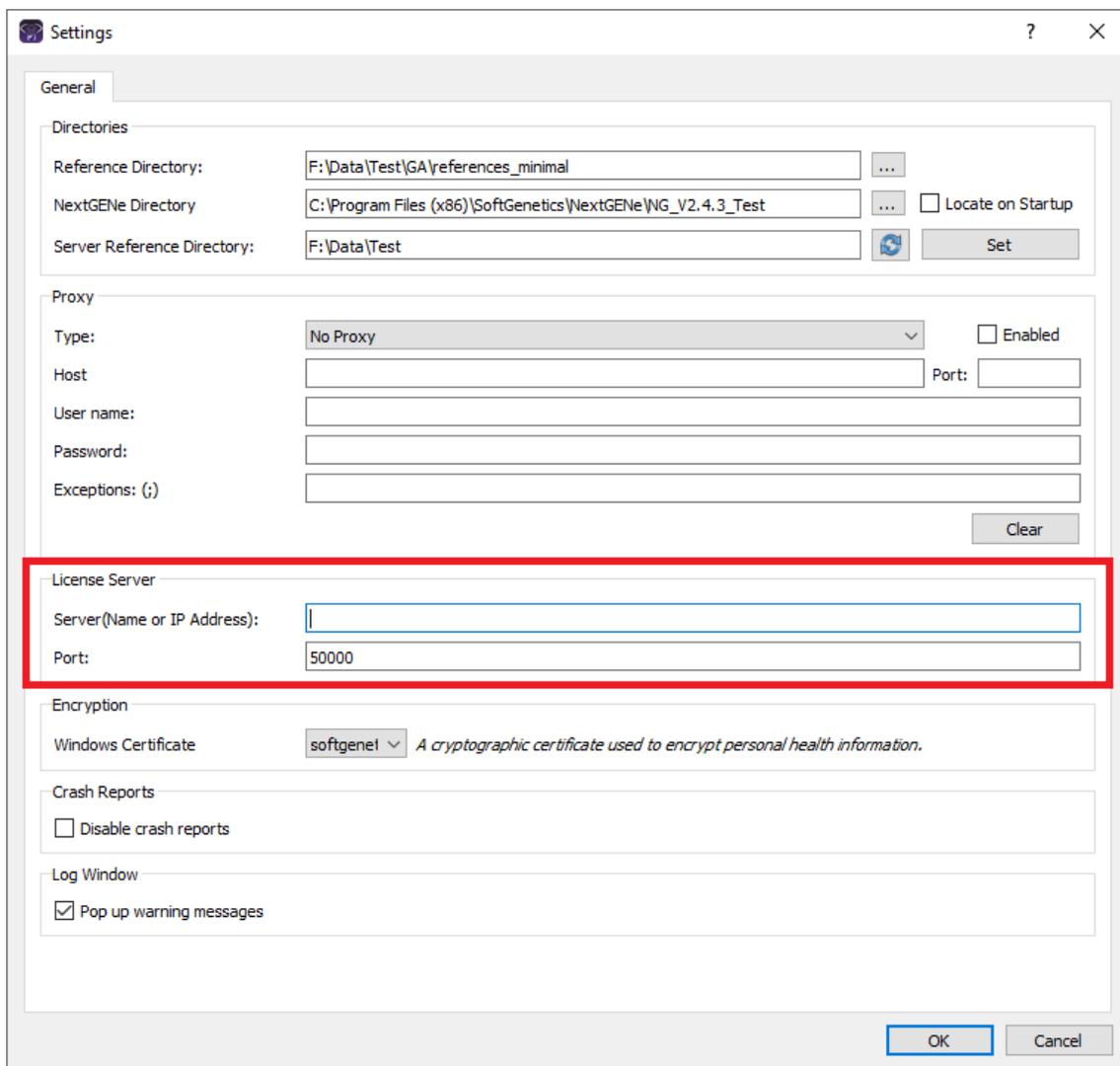
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• Configure License

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



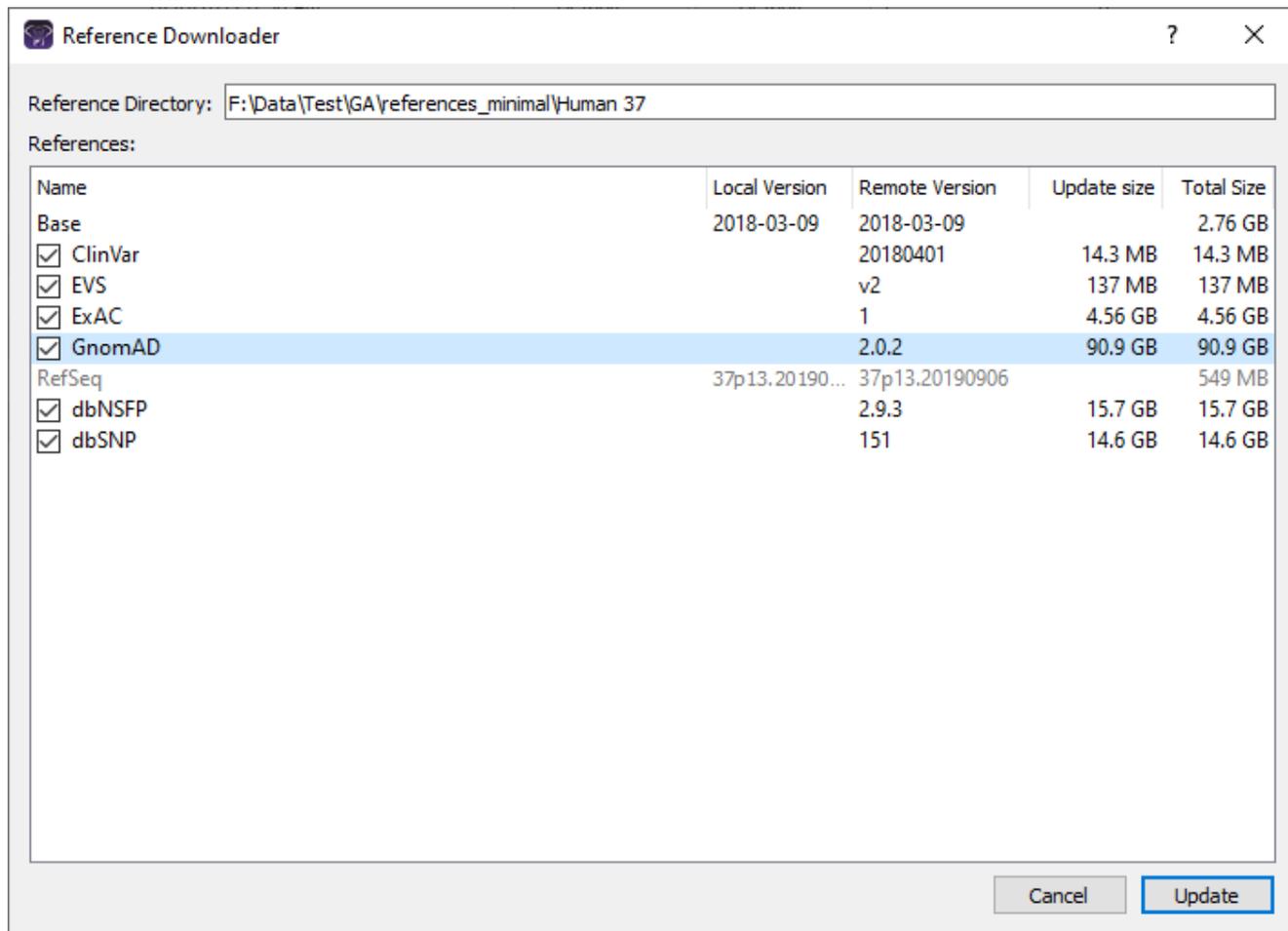
- Set the server to the computer with the License Server installed
 - The default port is 50000
 - License Server installation is described in a separate guide. Contact SoftGenetics to obtain this if needed.



Geneticist Assistant Quick Start Guide

• Download Annotation Databases

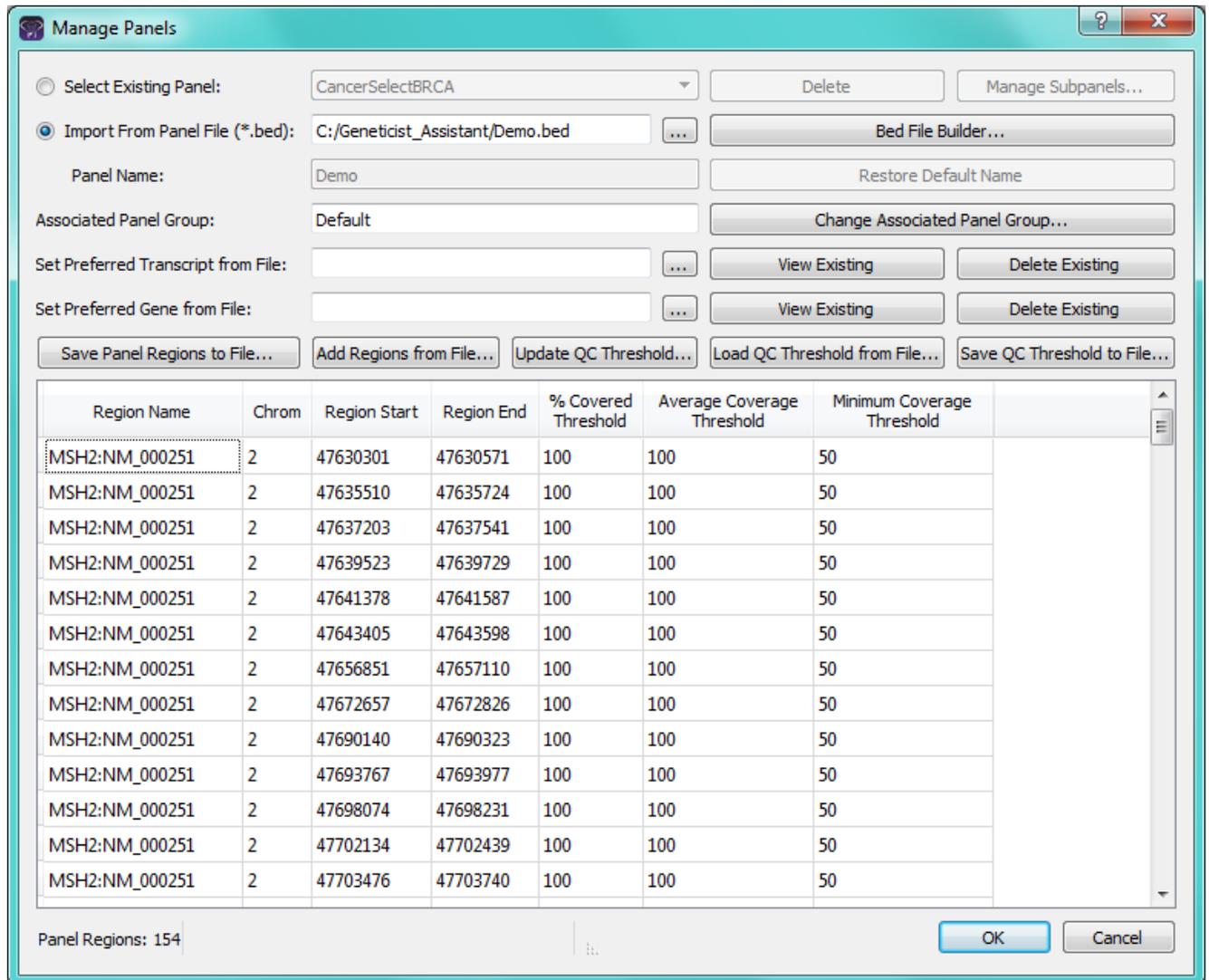
- Click the “Ref” button at the top right of Geneticist Assistant
- Select databases to be downloaded and click “Update”



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



Geneticist Assistant Quick Start Guide

• 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 New...

Instrument: Default New...

Reference: Human 37

Panel: Demo New...

Panel Group: Default

Quality Metric Profile: Demo New...

Owner: Administrator New...

Multiple Samples in VCF

Select Directory Select Variant and/or Coverage and/or Copy Number Variant Files...

Optional Settings:

Run Date/Time: 9/1/2020 2:03 PM

Sample Collection Date: Vcf File Bam File Select 9/1/2020 2:03 PM

User Group: Supervisor

Sample Group: New...

Submit Coverage: Subpanel:

Import from VCF Pathogenicity and Status Comment Artifact Type

Submit Hotspots

Accepted VCF filters: PASS Edit

Identify Patient ID

Add New Patient...

from Sample Name

from File

Split Sample Name by Separator: _ (underscore)

800402.igv-sorted_Output_Mutation_Report1_filtered.vcf:

Sample Name	Variant File(*.vcf)	Coverage/Pile Up File(*.bam)	Run Date/Time	Sample Collection Date	Sample Group	Submit Coverage
800402.igv-sorted_Output_Mutation_Report1_filtered.vcf	ssistant/Demo_data/800402.igv-sorted_Output_Mutation_Report1_filtered.vcf.gz	C:/Geneticist_Assistant/Demo_data/800402.igv-sorted_Output.bam	9/1/2020 2:07 PM	9/27/2016 1:54 PM		<input checked="" type="checkbox"/>

OK Cancel