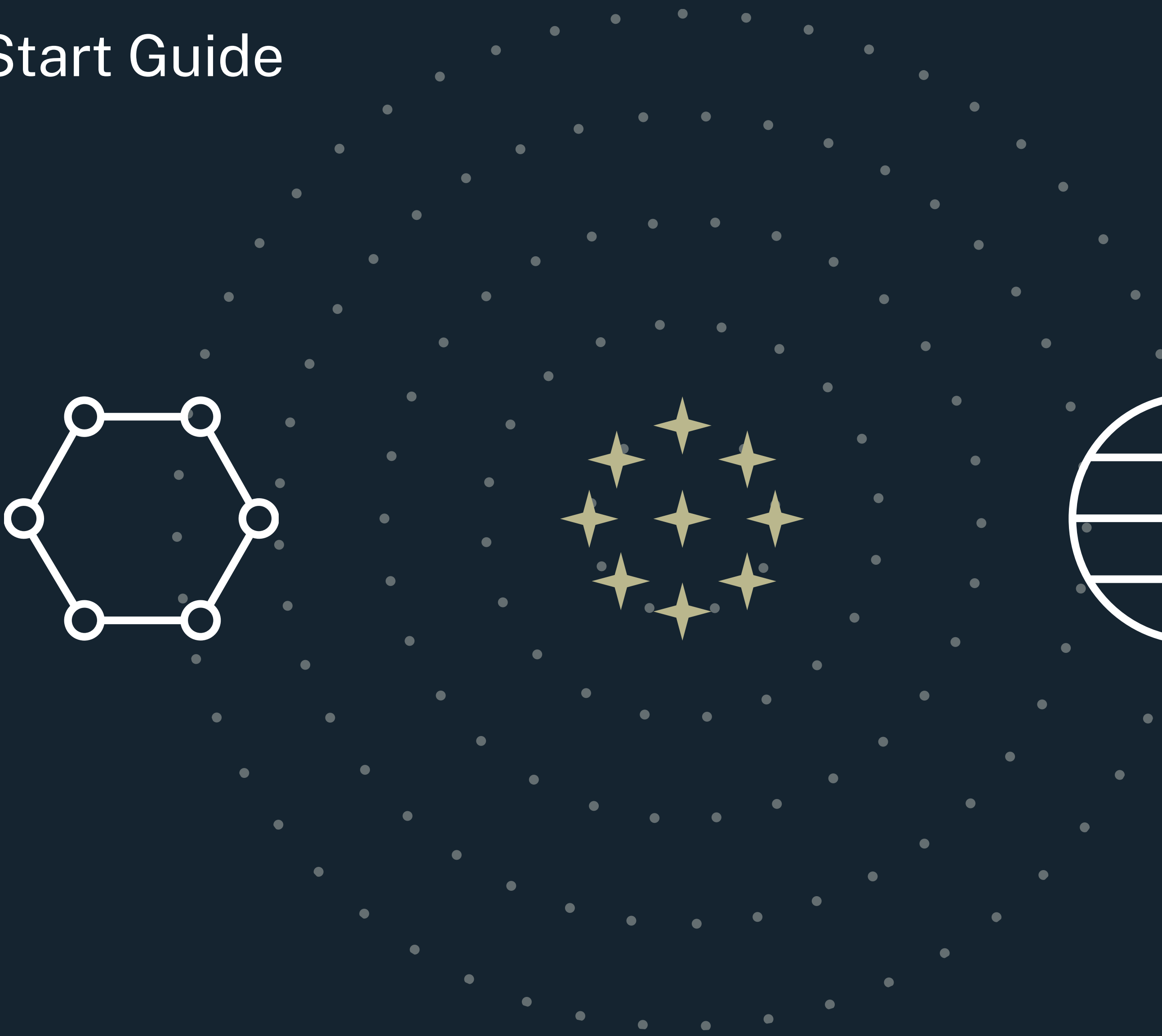
Dotmatics
SoftGenetics

Geneticist Assistant

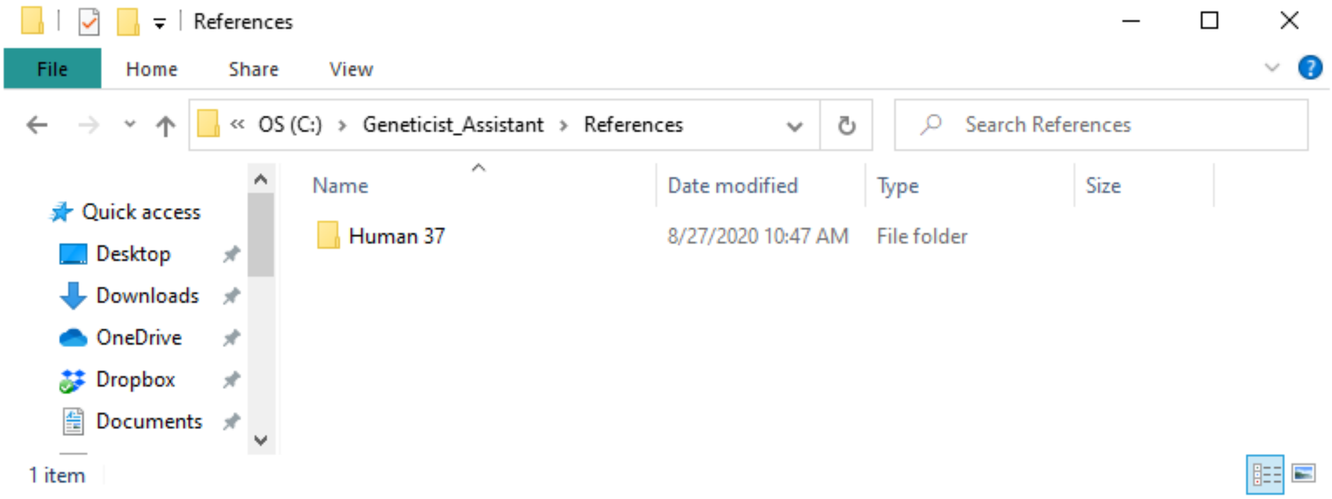
Quick Start Guide



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.

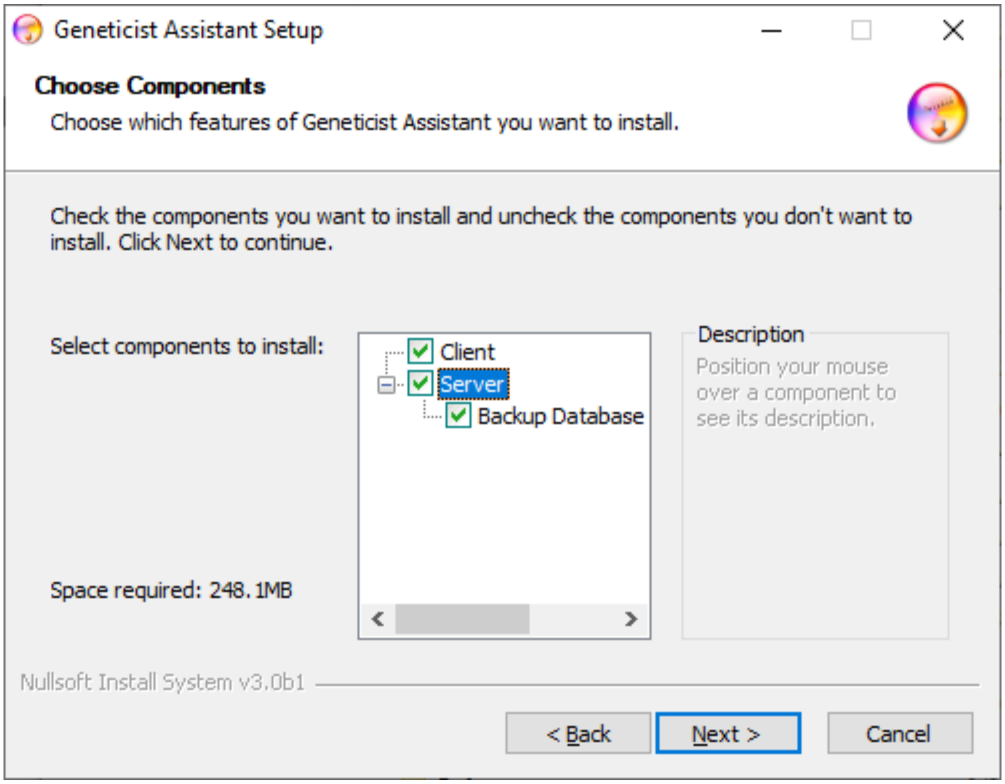


« OS (C:) › Geneticist_Assistant › References

Name	Date modified	Type	Size
Human 37	8/27/2020 10:47 AM	File folder	

Geneticist Assistant Quick Start Guide

- ## 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



Geneticist Assistant Setup

Choose Components

Choose which features of Geneticist Assistant you want to install.

Check the components you want to install and uncheck the components you don't want to install. Click Next to continue.

Select components to install:

- ☑ Client
- ☑ Server
  - ☑ Backup Database

Space required: 248.1MB

Description

Position your mouse over a component to see its description.

Nullsoft Install System v3.0b1

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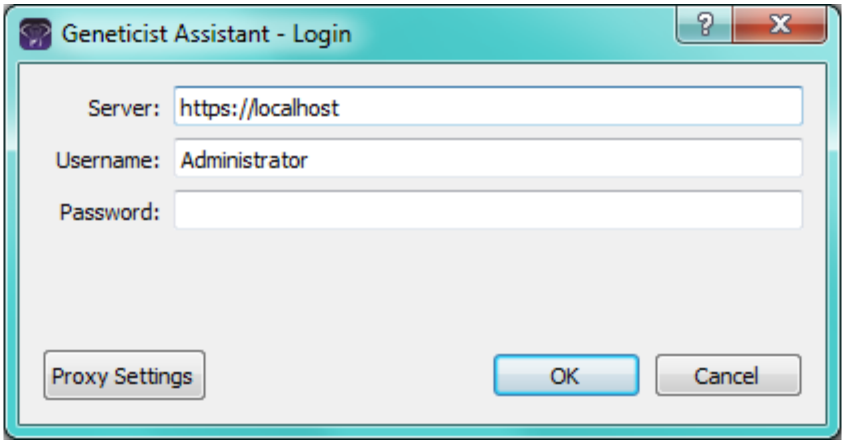
Log in

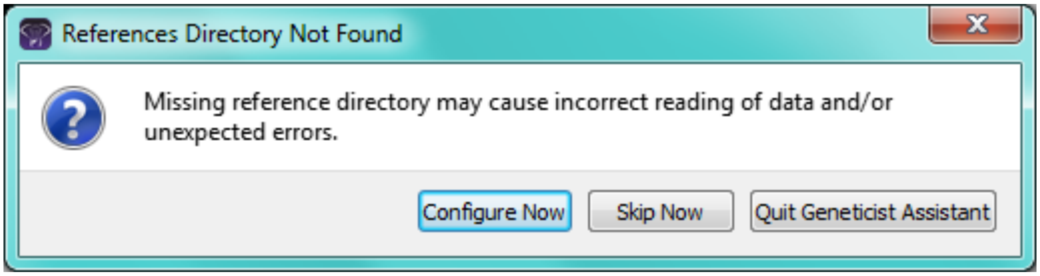
- Server: Name of the computer running the server
- Username: The user name you set up during install
- Password: The password you set up during install



Server: https://localhost
Username: Administrator
Password:

Configure Reference Directory

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



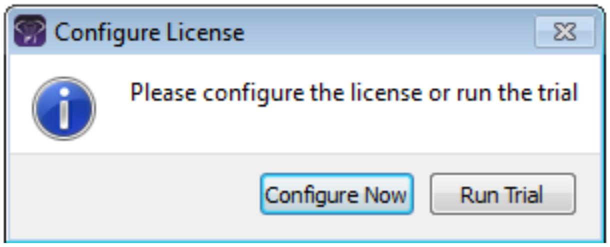
Missing reference directory may cause incorrect reading of data and/or unexpected errors.

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

Geneticist Assistant Quick Start Guide

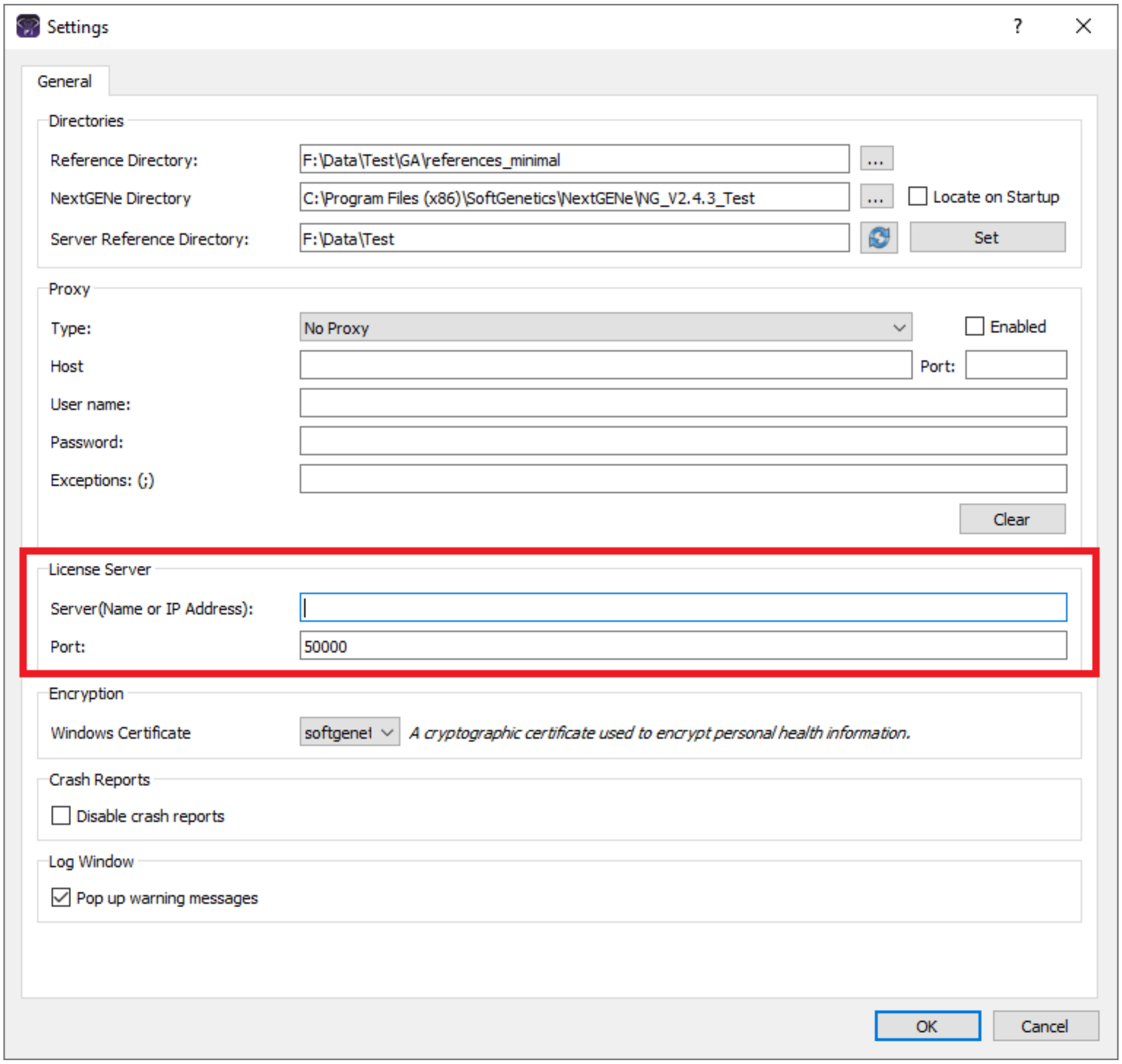
• Configure License

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



Please configure the license or run the 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.



Directories

Reference Directory: F:\Data\Test\GA\references_minimal

NextGENe Directory: C:\Program Files (x86)\SoftGenetics\NextGENe\NG_V2.4.3_Test ☐ Locate on Startup

Server Reference Directory: F:\Data\Test

Proxy

Type: No Proxy ☐ Enabled

Host Port:

User name:

Password:

Exceptions: (;)

License Server

Server(Name or IP Address):

Port: 50000

Encryption

Windows Certificate softgene *A cryptographic certificate used to encrypt personal health information.*

Crash Reports

☐ Disable crash reports

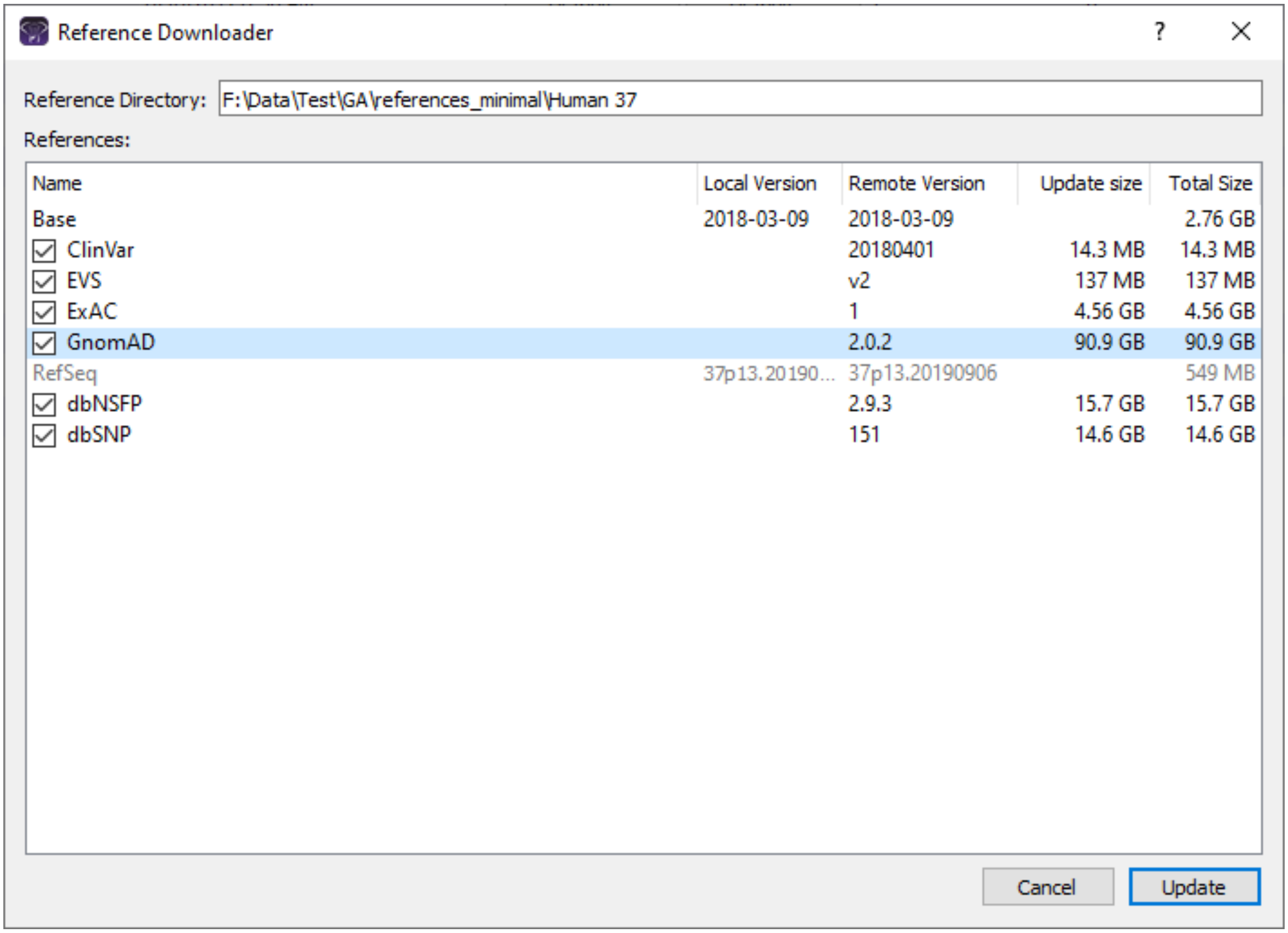
Log Window

☑ Pop up warning messages

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”



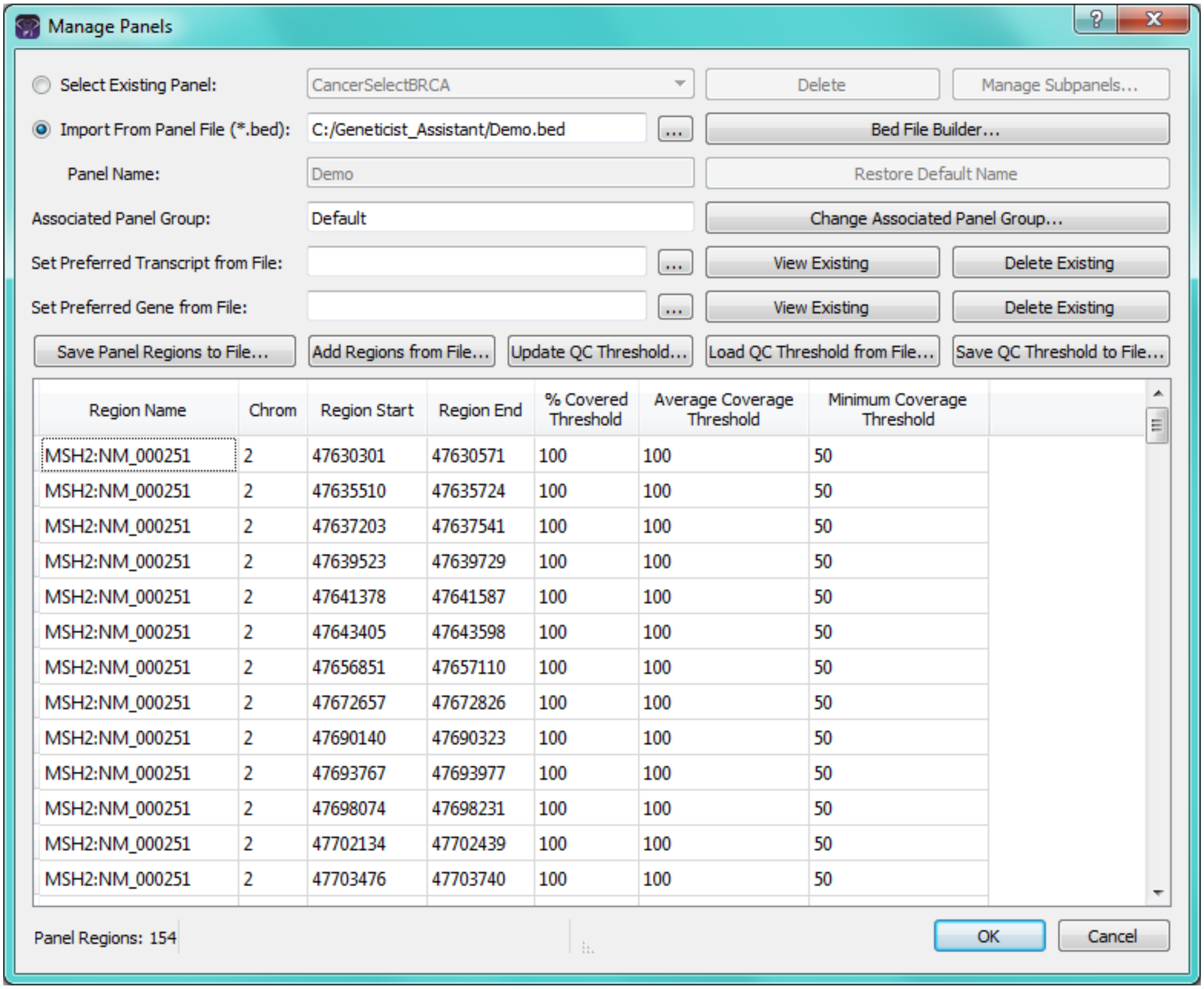
Reference Directory: F:\Data\Test\GA\references_minimal\Human 37

References:

Name	Local Version	Remote Version	Update size	Total Size
Base	2018-03-09	2018-03-09		2.76 GB
☑ ClinVar		20180401	14.3 MB	14.3 MB
☑ EVS		v2	137 MB	137 MB
☑ ExAC		1	4.56 GB	4.56 GB
☑ GnomAD		2.0.2	90.9 GB	90.9 GB
RefSeq	37p13.20190...	37p13.20190906		549 MB
☑ dbNSFP		2.9.3	15.7 GB	15.7 GB
☑ dbSNP		151	14.6 GB	14.6 GB

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



Manage Panels

○ Select Existing Panel: CancerSelectBRCA

● Import From Panel File (*.bed): C:/Geneticist_Assistant/Demo.bed

Panel Name: Demo

Associated Panel Group: Default

Set Preferred Transcript from File:

Set Preferred Gene from File:

Region Name	Chrom	Region Start	Region End	% Covered Threshold	Average Coverage Threshold	Minimum Coverage Threshold
MSH2:NM_000251	2	47630301	47630571	100	100	50
MSH2:NM_000251	2	47635510	47635724	100	100	50
MSH2:NM_000251	2	47637203	47637541	100	100	50
MSH2:NM_000251	2	47639523	47639729	100	100	50
MSH2:NM_000251	2	47641378	47641587	100	100	50
MSH2:NM_000251	2	47643405	47643598	100	100	50
MSH2:NM_000251	2	47656851	47657110	100	100	50
MSH2:NM_000251	2	47672657	47672826	100	100	50
MSH2:NM_000251	2	47690140	47690323	100	100	50
MSH2:NM_000251	2	47693767	47693977	100	100	50
MSH2:NM_000251	2	47698074	47698231	100	100	50
MSH2:NM_000251	2	47702134	47702439	100	100	50
MSH2:NM_000251	2	47703476	47703740	100	100	50

Panel Regions: 154

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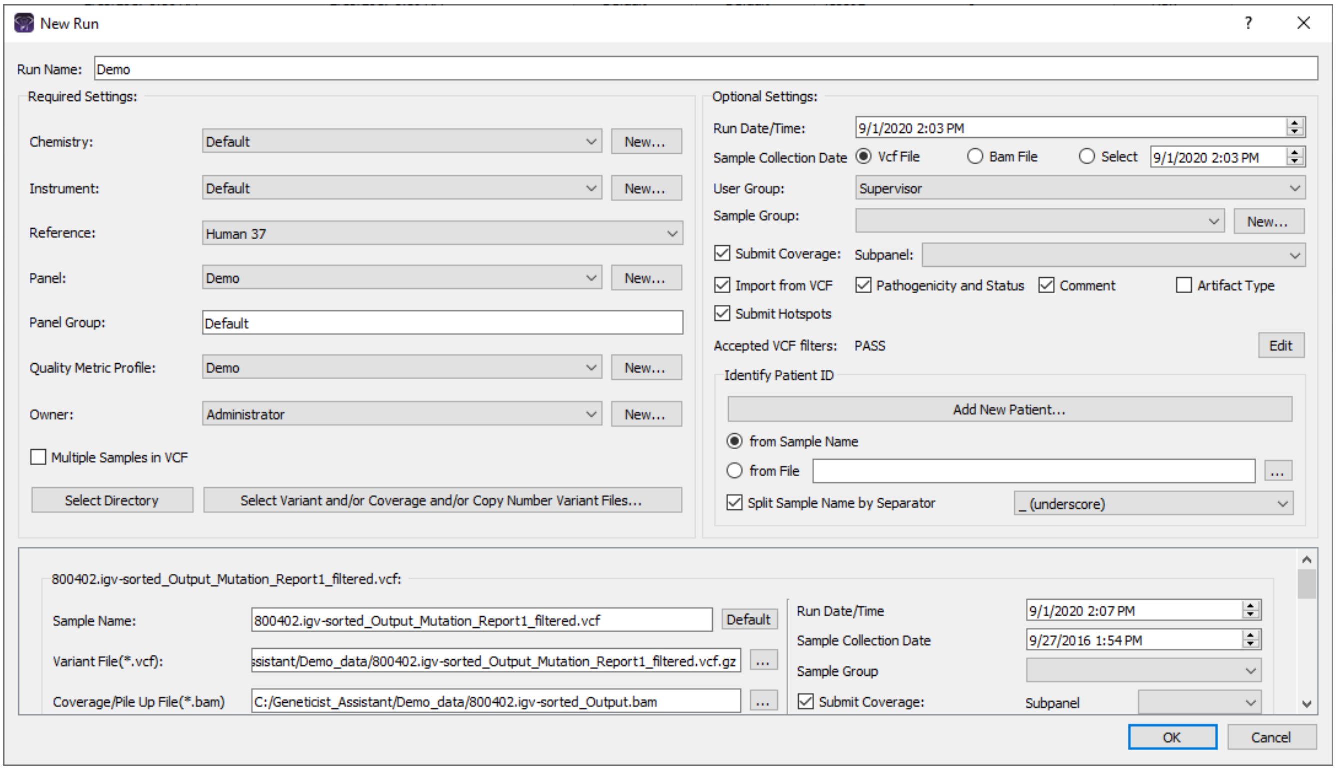
Submit New Run

- 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.



Run Name: Demo

Required Settings:

Chemistry: Default

Instrument: Default

Reference: Human 37

Panel: Demo

Panel Group: Default

Quality Metric Profile: Demo

Owner: Administrator

☐ Multiple Samples in VCF

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:

☑ Submit Coverage: Subpanel:

☑ Import from VCF ☑ Pathogenicity and Status ☑ Comment ☐ Artifact Type

☑ Submit Hotspots

Accepted VCF filters: PASS

Identify Patient ID

● from Sample Name

○ from File

☑ Split Sample Name by Separator _ (underscore)

800402.igv-sorted_Output_Mutation_Report1_filtered.vcf:

Sample Name: 800402.igv-sorted_Output_Mutation_Report1_filtered.vcf

Variant File(*.vcf): ssistant/Demo_data/800402.igv-sorted_Output_Mutation_Report1_filtered.vcf.gz

Coverage/Pile Up File(*.bam) C:/Geneticist_Assistant/Demo_data/800402.igv-sorted_Output.bam

Run Date/Time 9/1/2020 2:07 PM

Sample Collection Date 9/27/2016 1:54 PM

Sample Group

☑ Submit Coverage: Subpanel