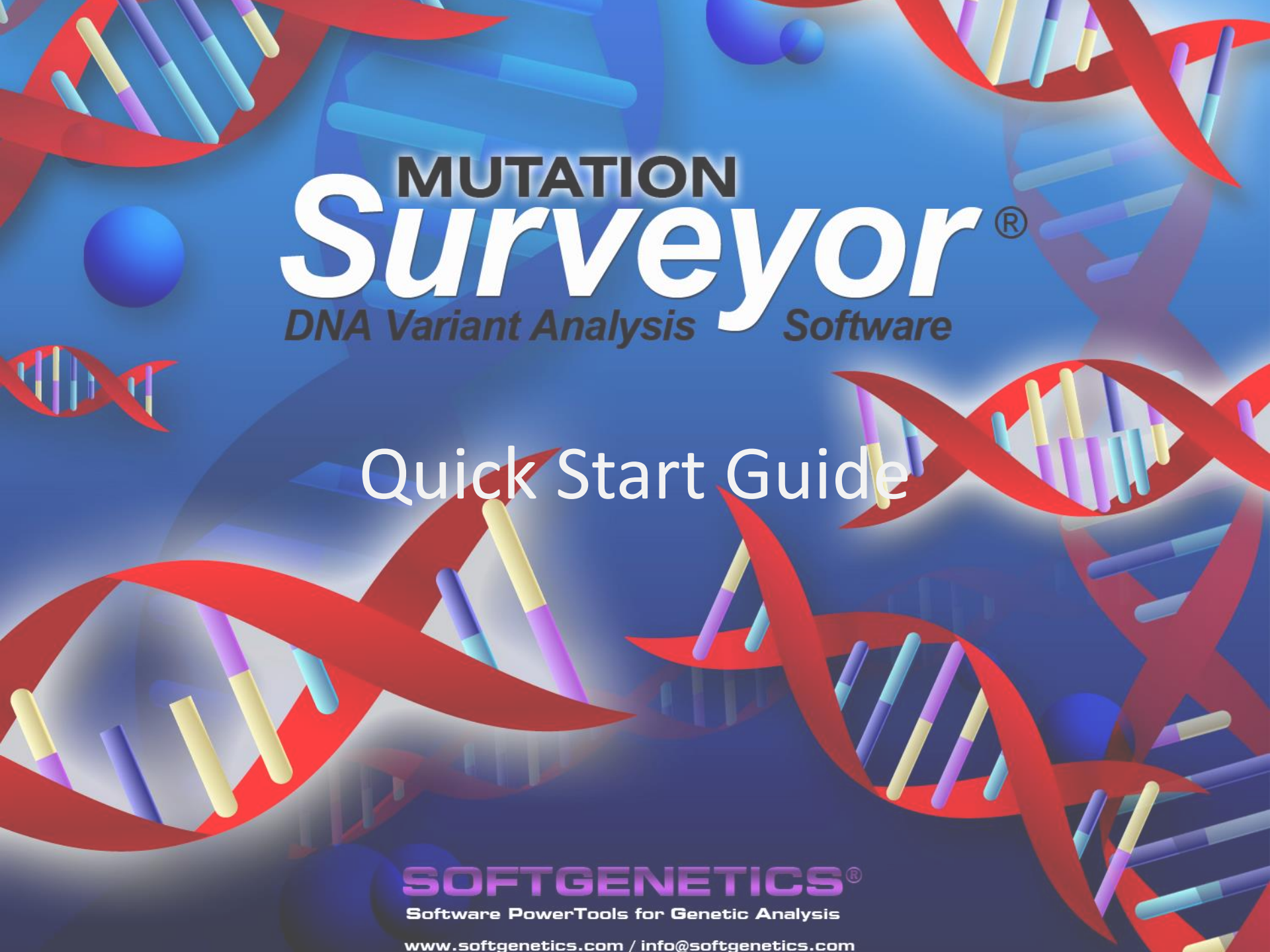
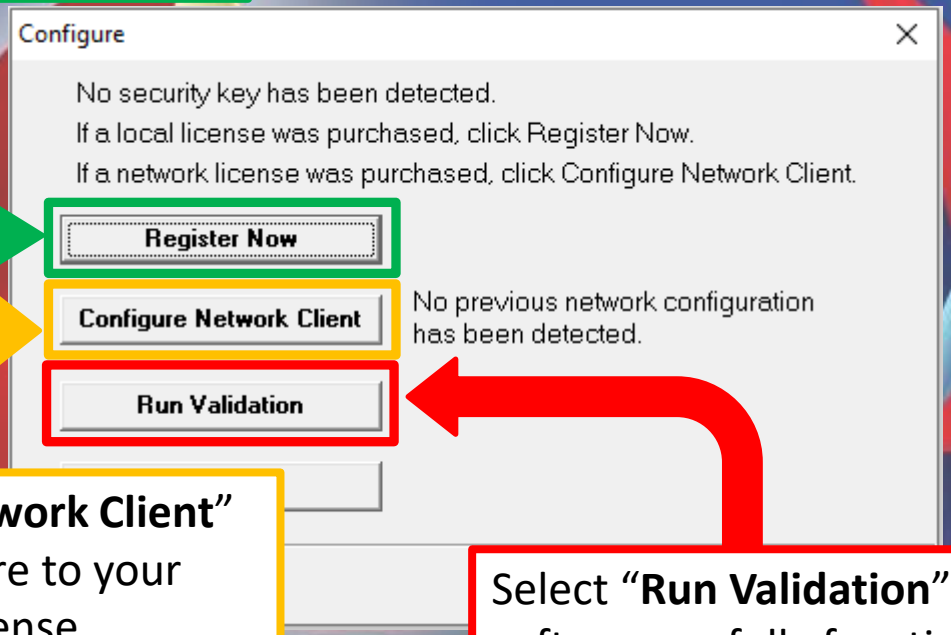

MUTATION
Surveyor®
DNA Variant Analysis Software
Quick Start Guide
SOFTGENETICS®
Software PowerTools for Genetic Analysis
www.softgenetics.com / info@softgenetics.com

Opening Software for the First Time

Select "**Register Now**" to register your purchased local license

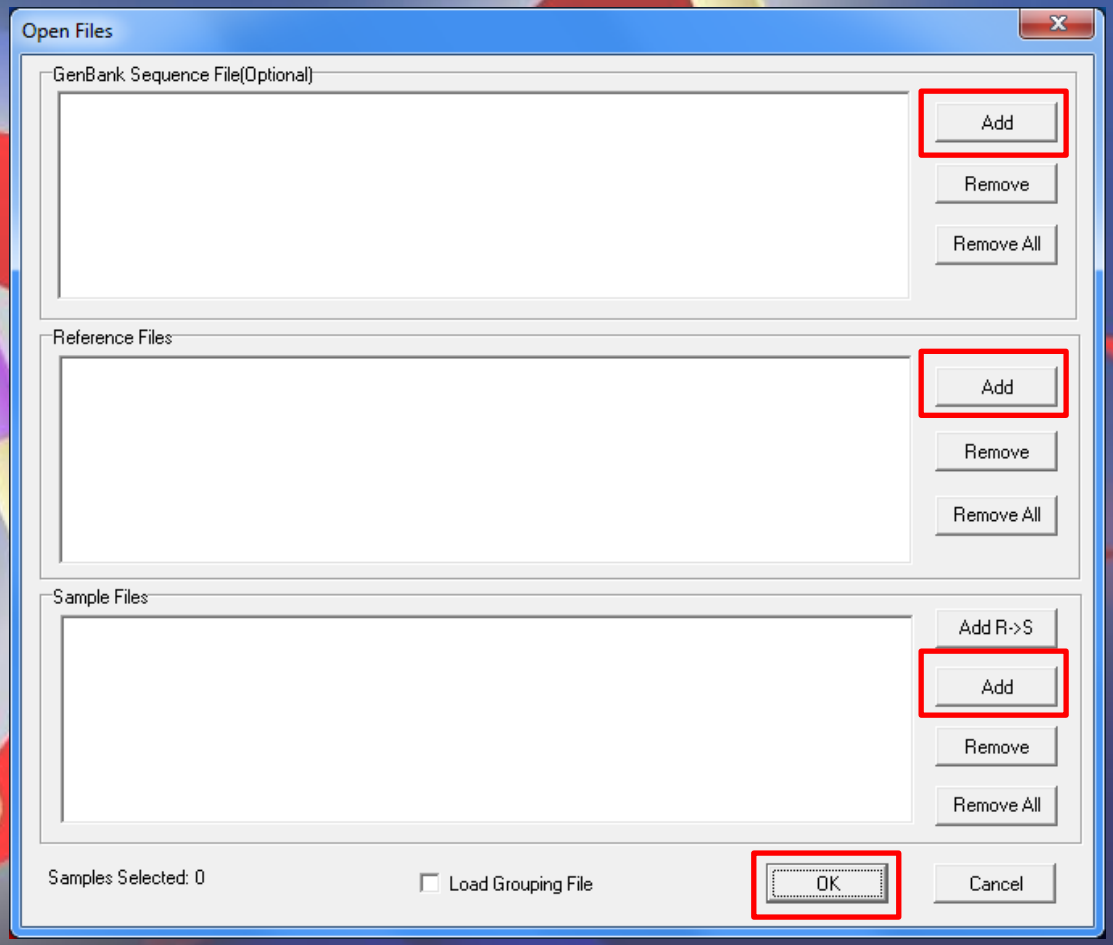


Select "**Configure Network Client**" to connect the software to your registered network license

Select "**Run Validation**" to open software as fully functional trial (trial will expire 35 days from the time this is first selected)

Upload Data

Click **Open Files** icon in the main toolbar



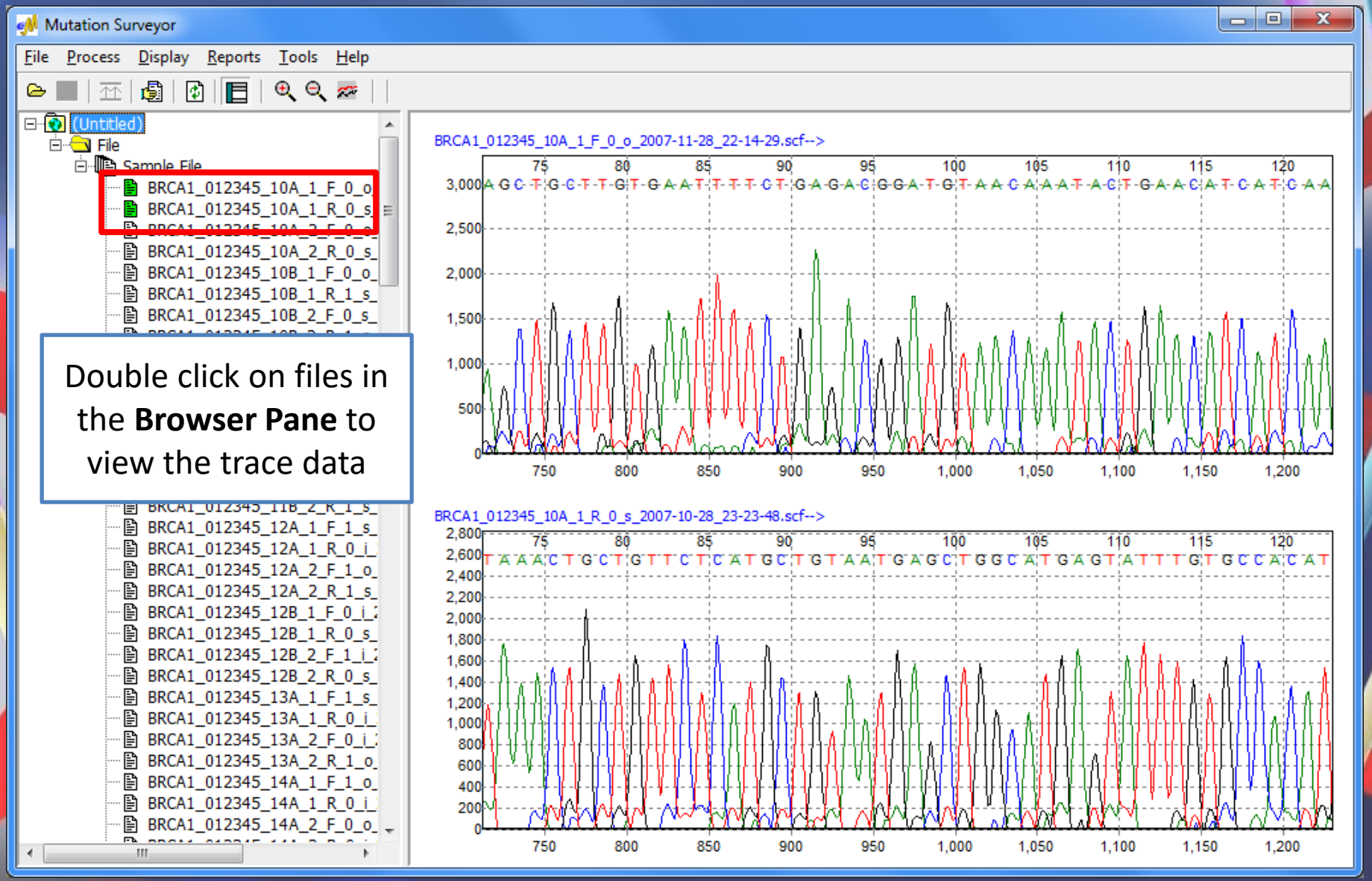
Add **GenBank Files** (GB/GBK/SEQ)
and/or
Reference Trace Files (SCF/AB1/ABI)

NOTE: Both GenBank and Reference files are optional when sequence maps to genes of *Homo sapiens*.

Add **Sample Files** (SCF/AB1/ABI)

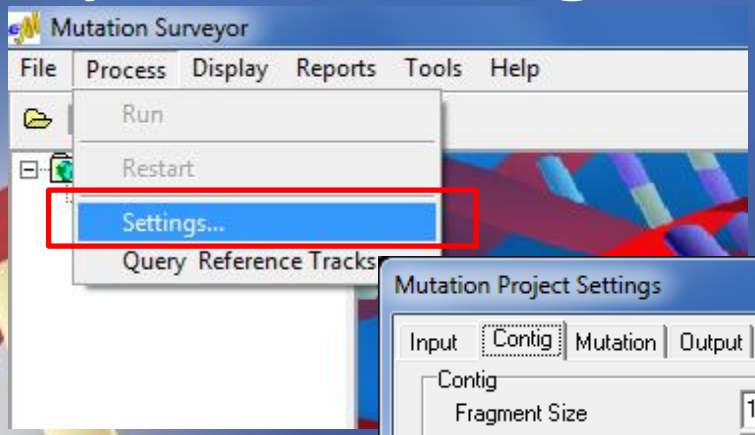
Click OK

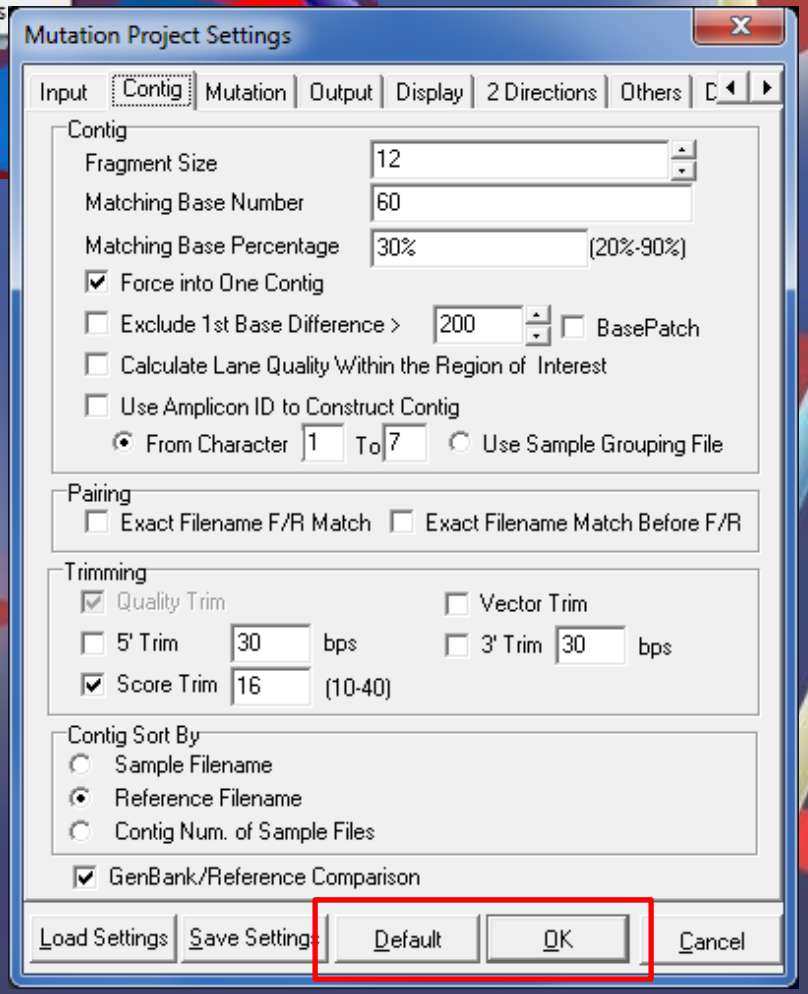
Review Raw Data



Analysis Settings

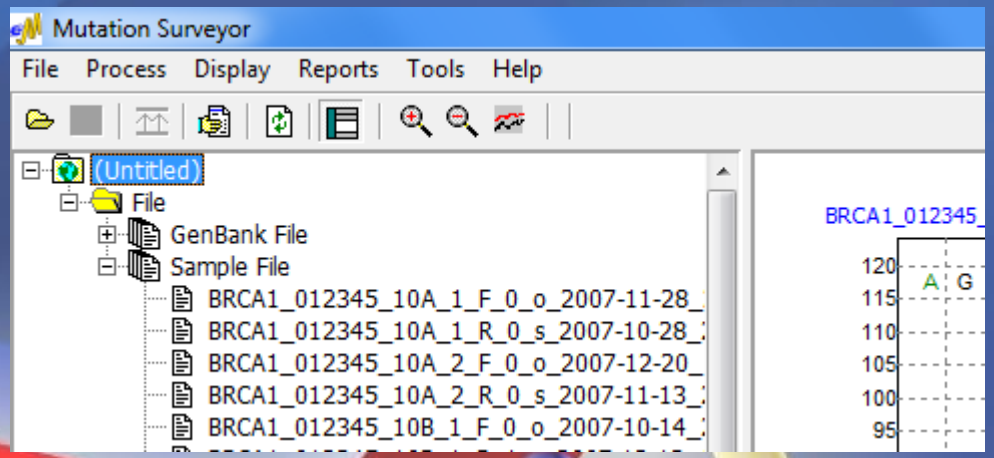
In the main toolbar select **Process → Settings**





For first run analysis select **Default** and then select **OK**

Analyze Data



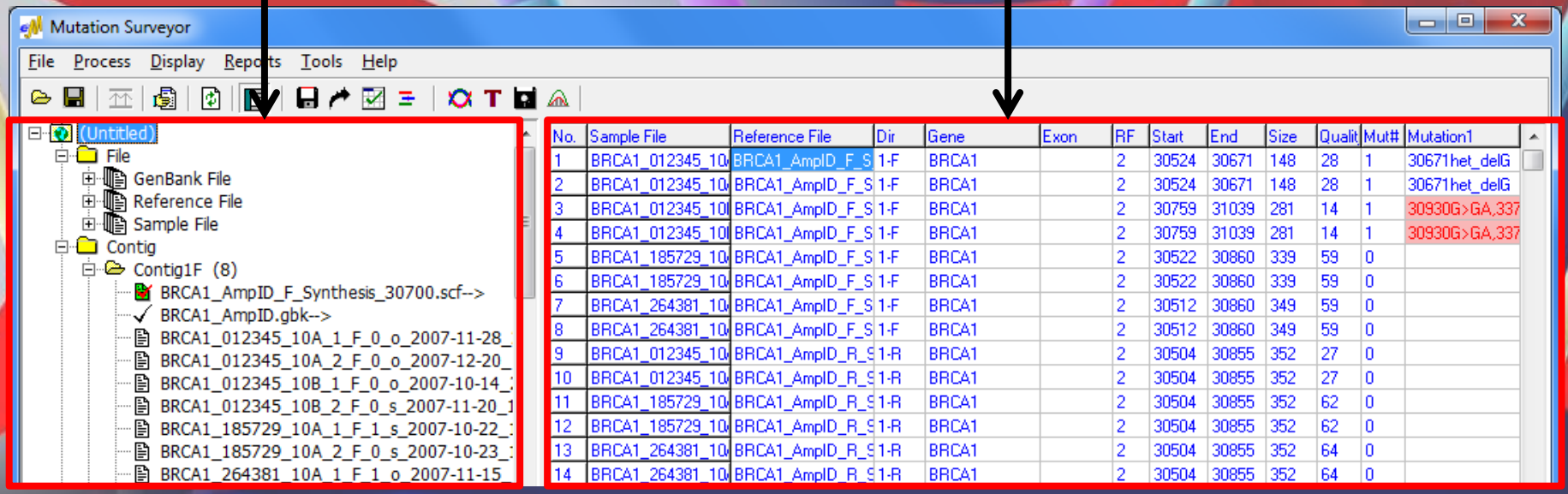
Click **Run** icon in main toolbar to begin analysis

Your samples will be compared to references

The first window to appear is the **Mutation Report**

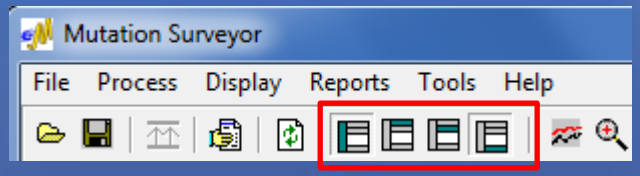
Browser Pane

Mutation Report



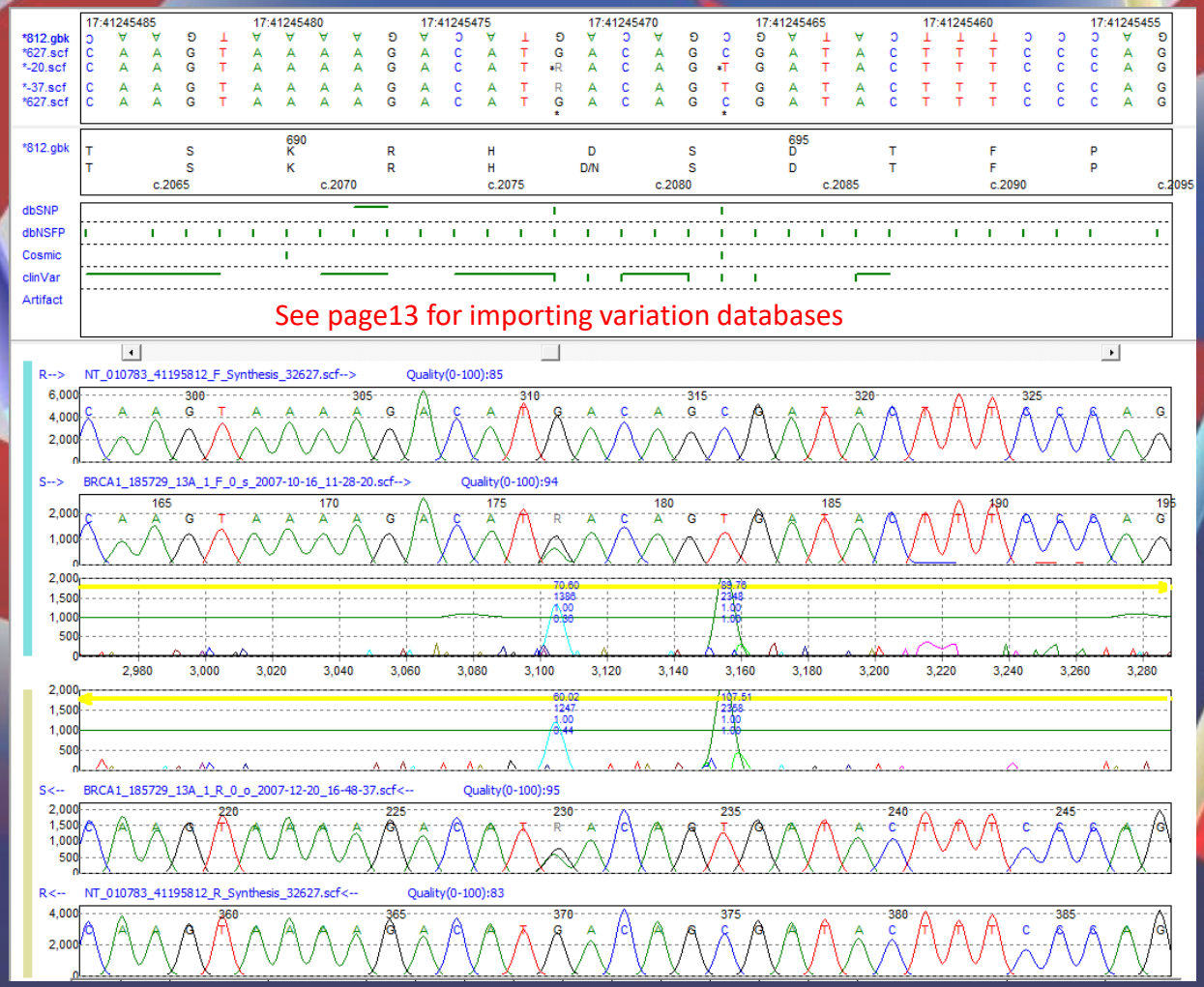
Graphic Analysis Display

Use icons to show/hide windows

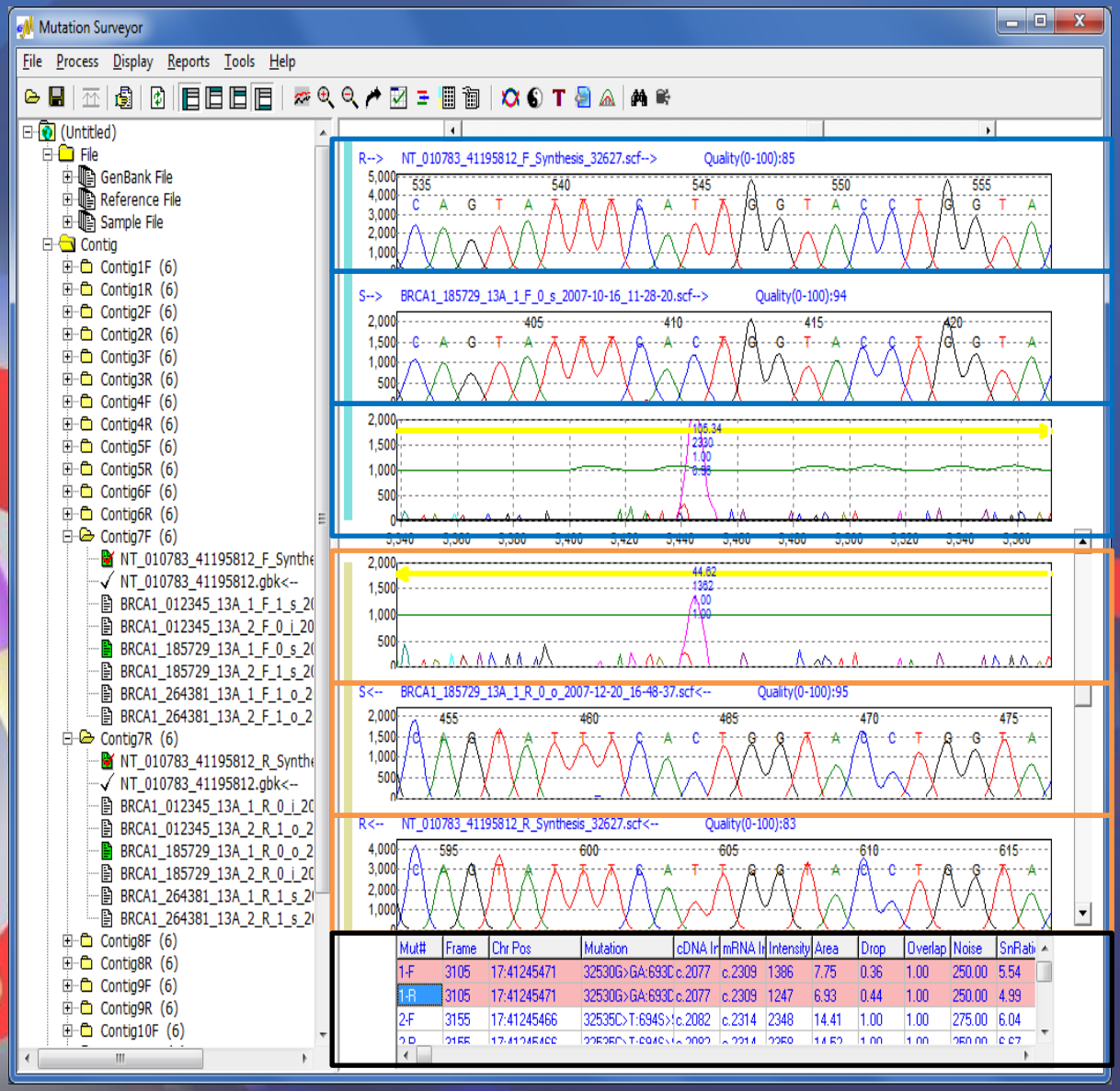


Nucleotide, **Amino Acid**, and **Database** window

Trace windows



Graphic Analysis Display



By double clicking a mutation cell in the **Mutation Report**, the **Graphic Analysis Display** window appears

Forward Reference Trace

Forward Sample Trace

Forward Comparison

Reverse Comparison

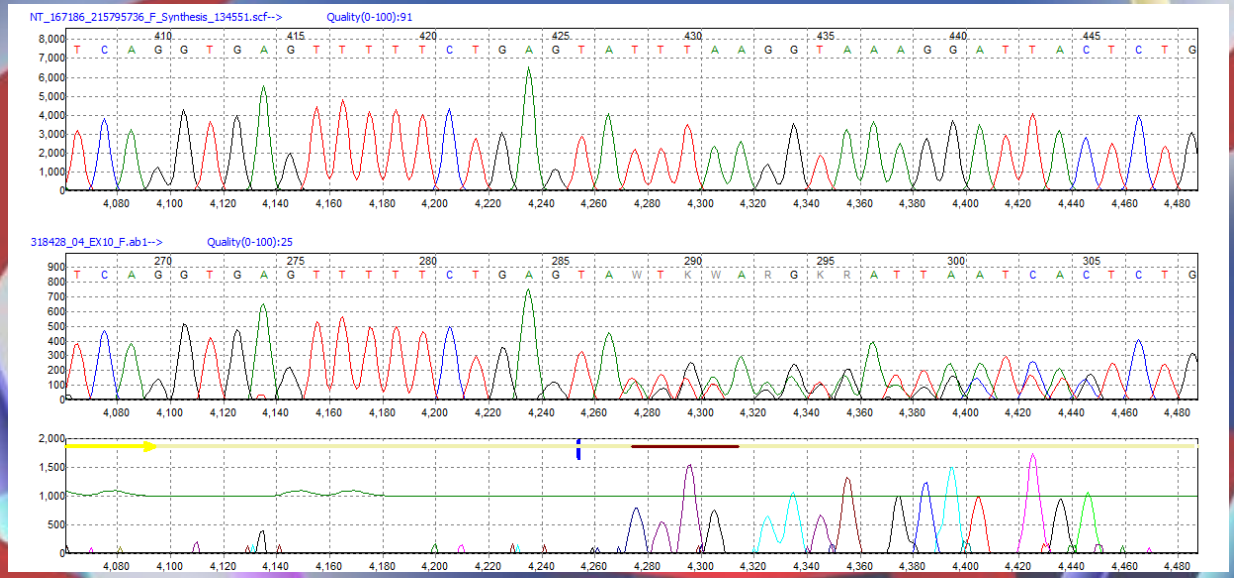
Reverse Sample Trace

Reverse Reference Trace

Mutation Table

Heterozygous Indel Detection

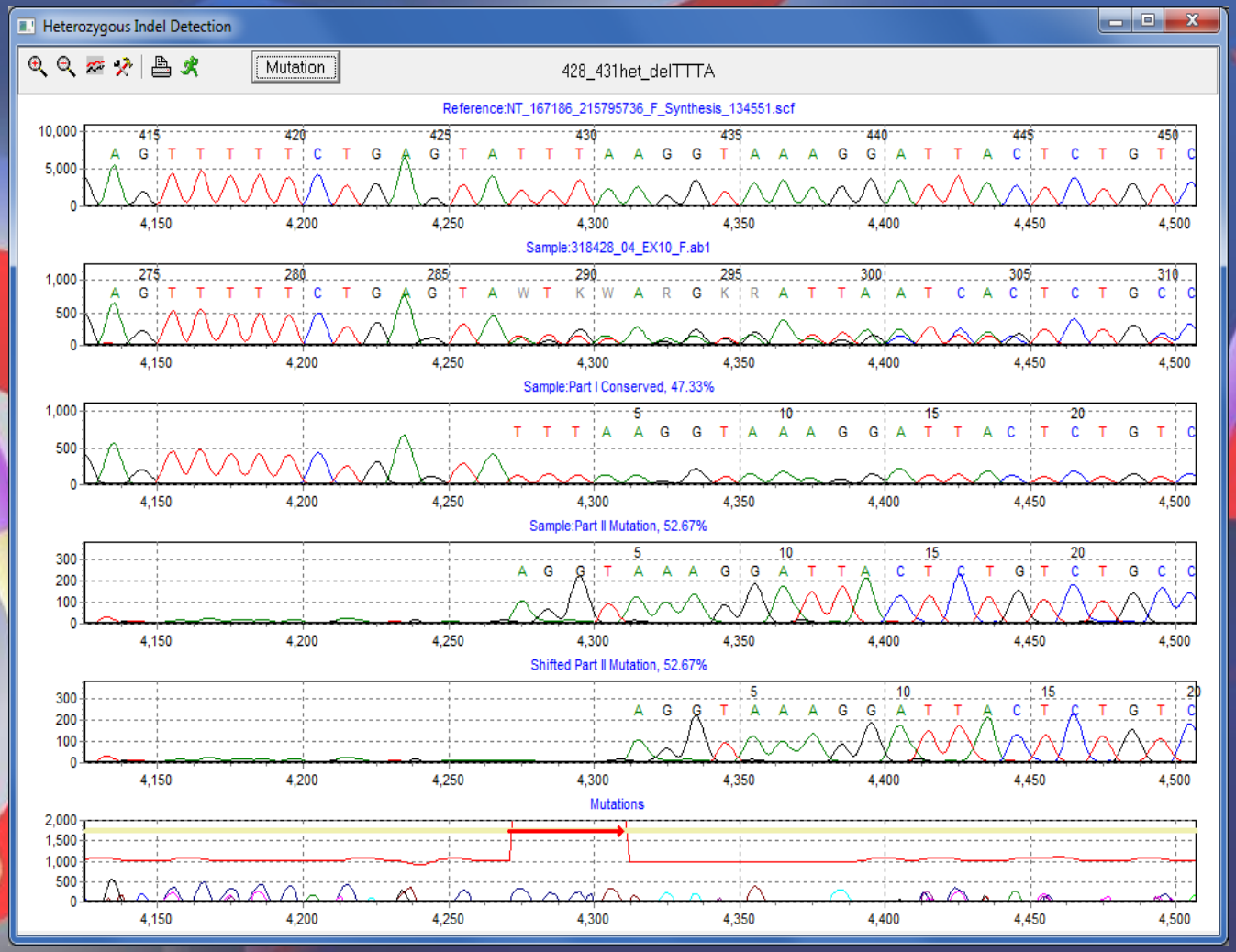
Brown bar in Mutation Electropherogram indicates that a heterozygous indel is detected



Double click mutation call or select icon to open heterozygous indel deconvolution tool

18	465130_10_EX10_F	NT_167186_215795	1-F	USH2A	11	2	134375	134750	376	38	1	
19	465130_12_EX10_F	NT_167186_215795	1-F	USH2A	11	2	134365	134534	170	0	1	134533_134535het_dupTGC

Heterozygous Indel Deconvolution Tool



Reference Trace

Sample Trace

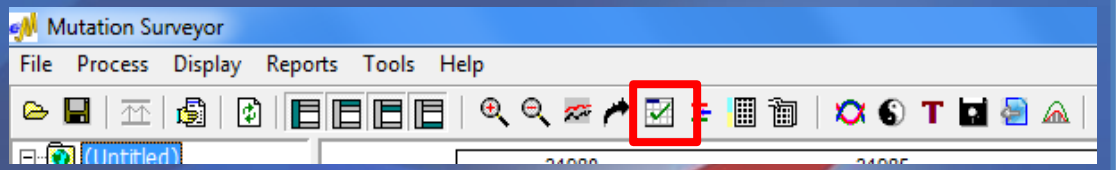
Reference/Wild Type Allele

Mutant/Novel Allele

Mutant Allele Re-aligned to Reference

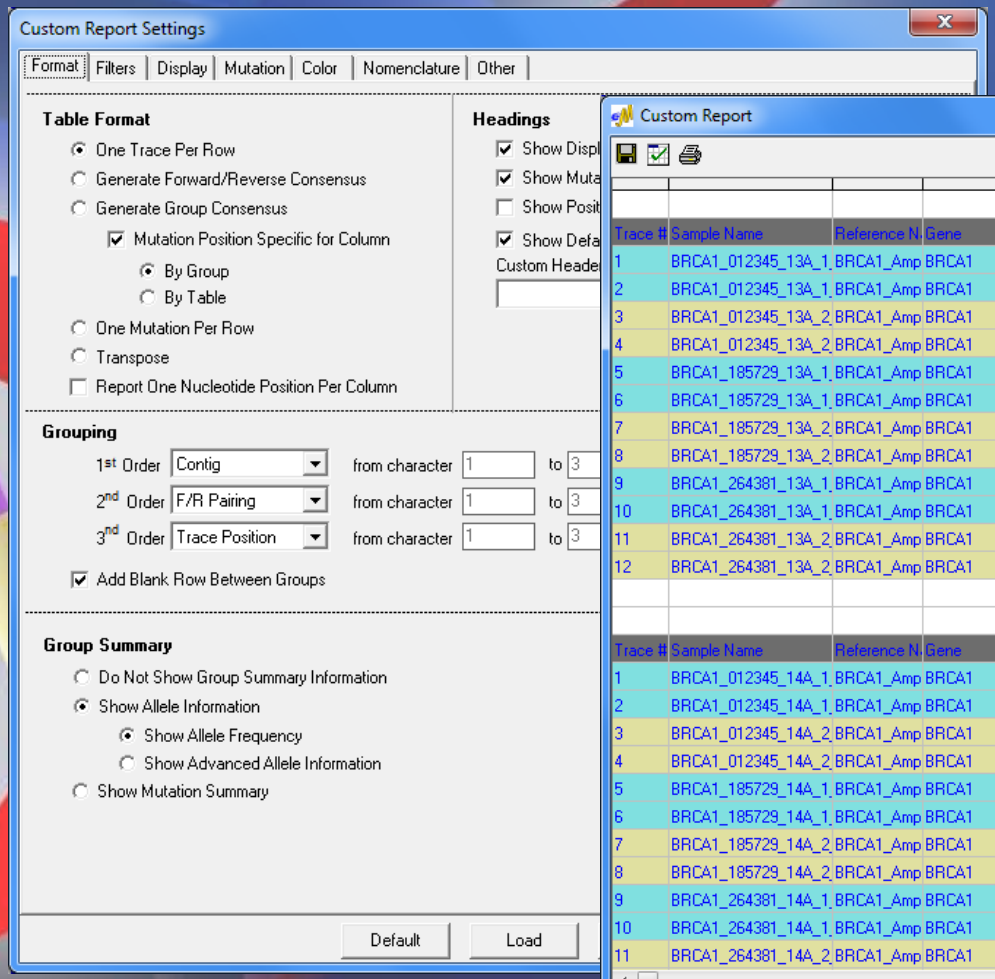
Location of Indel

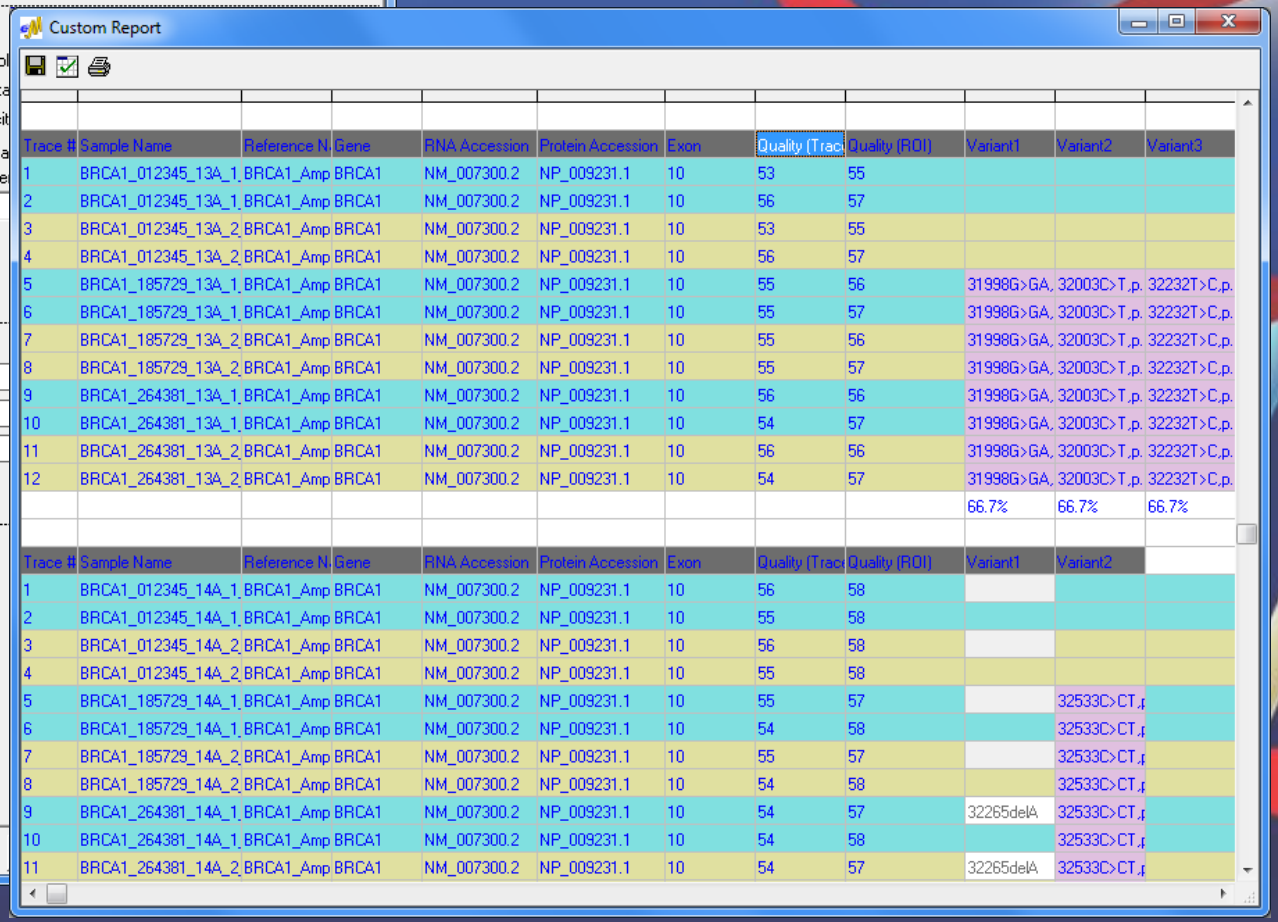
Custom Report Builder



Click the **Custom Report Builder** Icon in the main toolbar

Default settings group by Contig and pair Forward and Reverse files

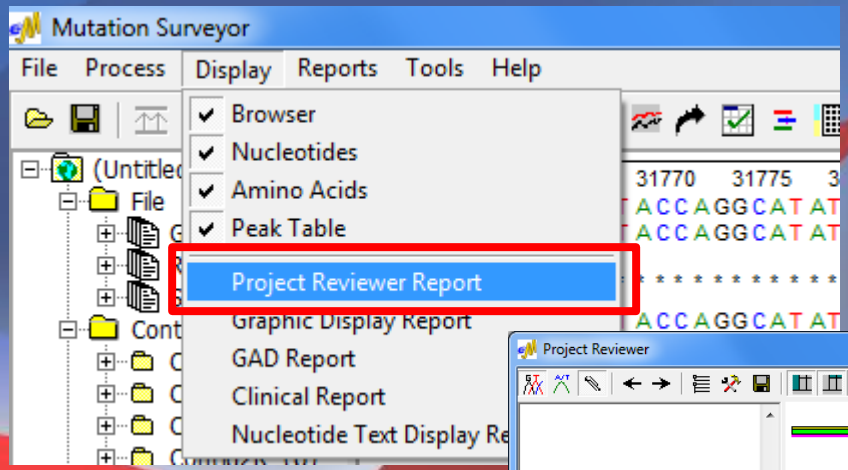




Trace #	Sample Name	Reference N.	Gene	RNA Accession	Protein Accession	Exon	Quality (Trac	Quality (ROI)	Variant1	Variant2	Variant3
1	BRCA1_012345_13A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	53	55			
2	BRCA1_012345_13A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	56	57			
3	BRCA1_012345_13A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	53	55			
4	BRCA1_012345_13A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	56	57			
5	BRCA1_185729_13A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	56	31998G>GA,	32003C>T,p.	32232T>C,p.
6	BRCA1_185729_13A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	57	31998G>GA,	32003C>T,p.	32232T>C,p.
7	BRCA1_185729_13A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	56	31998G>GA,	32003C>T,p.	32232T>C,p.
8	BRCA1_185729_13A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	57	31998G>GA,	32003C>T,p.	32232T>C,p.
9	BRCA1_264381_13A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	56	56	31998G>GA,	32003C>T,p.	32232T>C,p.
10	BRCA1_264381_13A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	57	31998G>GA,	32003C>T,p.	32232T>C,p.
11	BRCA1_264381_13A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	56	56	31998G>GA,	32003C>T,p.	32232T>C,p.
12	BRCA1_264381_13A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	57	31998G>GA,	32003C>T,p.	32232T>C,p.
									66.7%	66.7%	66.7%

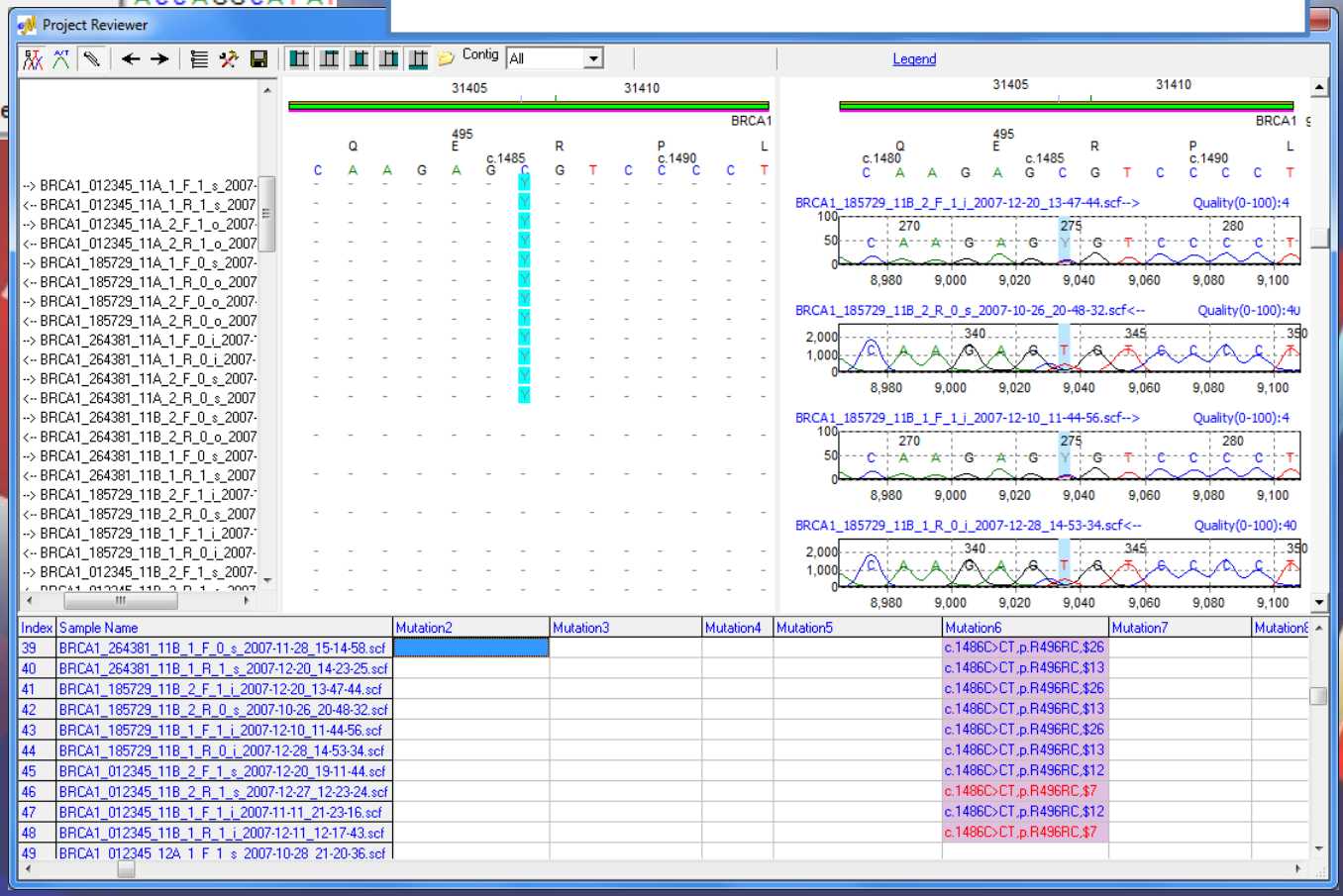
Trace #	Sample Name	Reference N.	Gene	RNA Accession	Protein Accession	Exon	Quality (Trac	Quality (ROI)	Variant1	Variant2	Variant3
1	BRCA1_012345_14A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	56	58			
2	BRCA1_012345_14A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	58			
3	BRCA1_012345_14A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	56	58			
4	BRCA1_012345_14A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	58			
5	BRCA1_185729_14A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	57		32533C>CT,p	
6	BRCA1_185729_14A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	58		32533C>CT,p	
7	BRCA1_185729_14A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	55	57		32533C>CT,p	
8	BRCA1_185729_14A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	58		32533C>CT,p	
9	BRCA1_264381_14A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	57	32265delA	32533C>CT,p	
10	BRCA1_264381_14A_1	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	58		32533C>CT,p	
11	BRCA1_264381_14A_2	BRCA1_Amp	BRCA1	NM_007300.2	NP_009231.1	10	54	57	32265delA	32533C>CT,p	

Project Reviewer

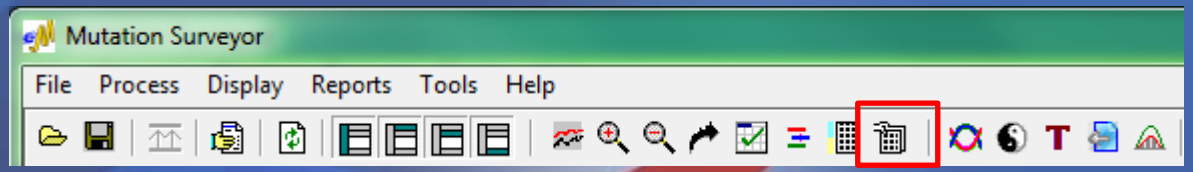


Click **Display** menu in main toolbar and choose **Project Reviewer Report**

Show all contigs simultaneously, group traces by sample ID, view information in four panes

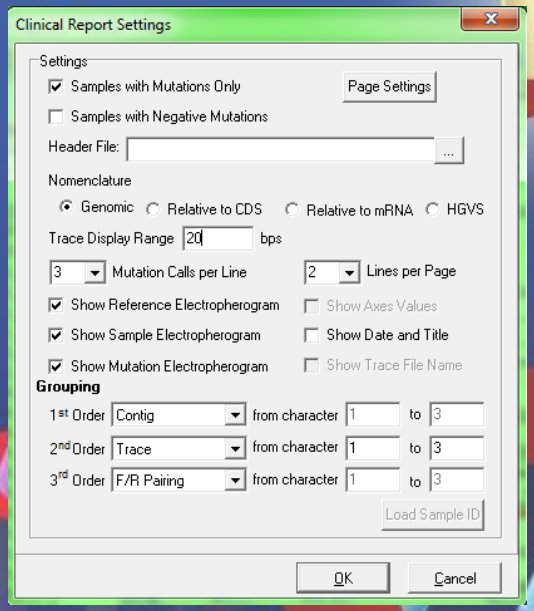


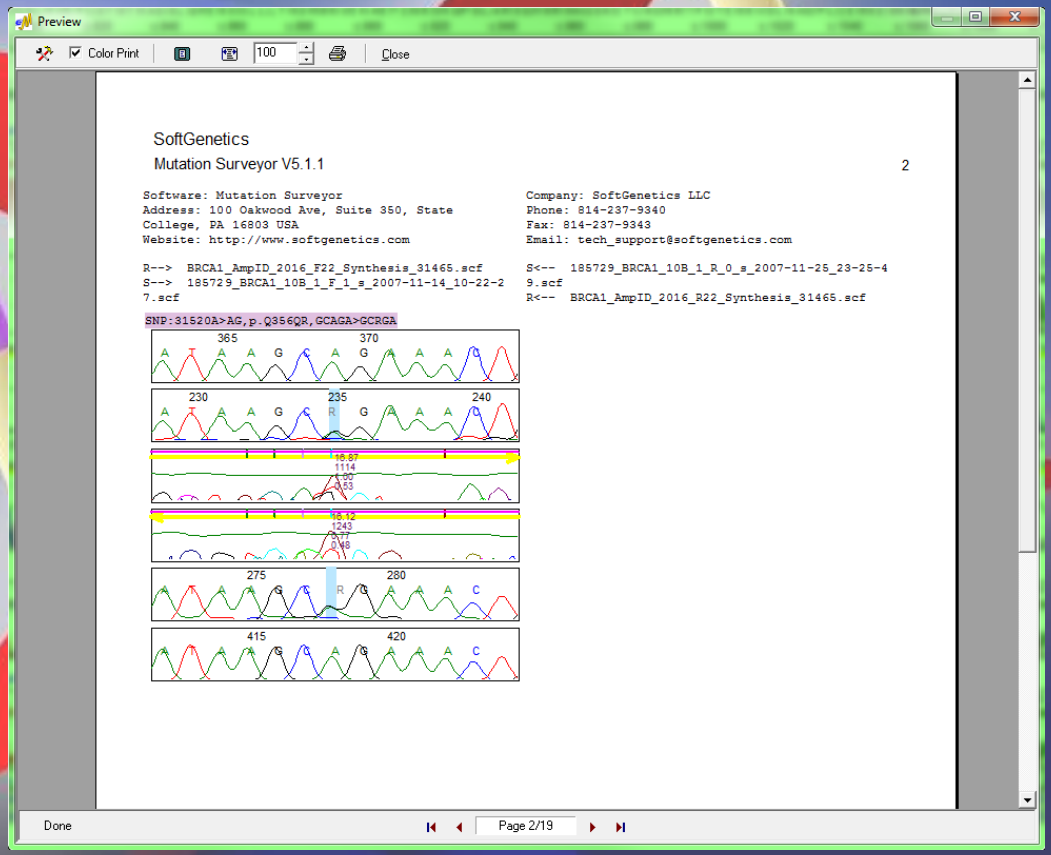
Print Clinical Report



Click **Clinical Report** Icon in main toolbar

Adjust Display preferences
Add a **Custom Header** file

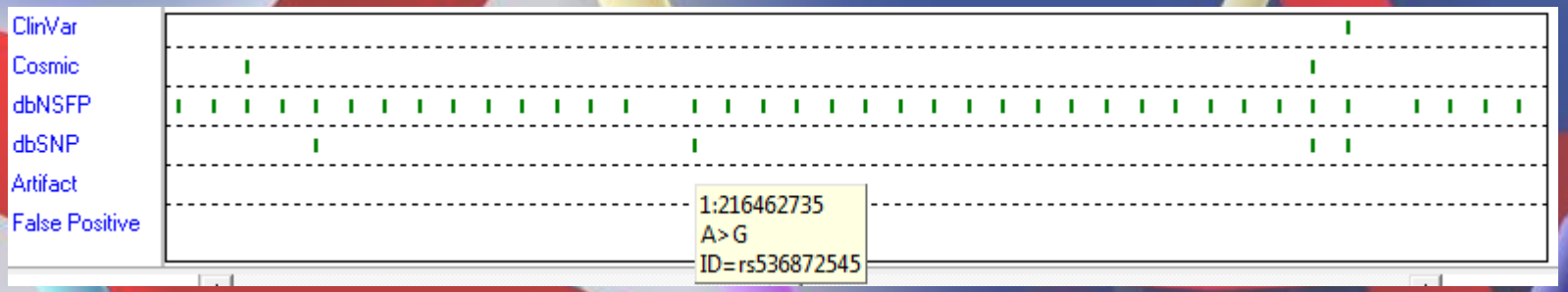




View and Print a snapshot of each mutation call

Variation Databases

Variant annotation from external databases can be imported and displayed in the Variation Tracks pane of the Graphical Analysis Display. The Tracks pane will also display the positions of any known deleted mutation calls that have been added to the User Knowledge Database as either a false positive or artifact.

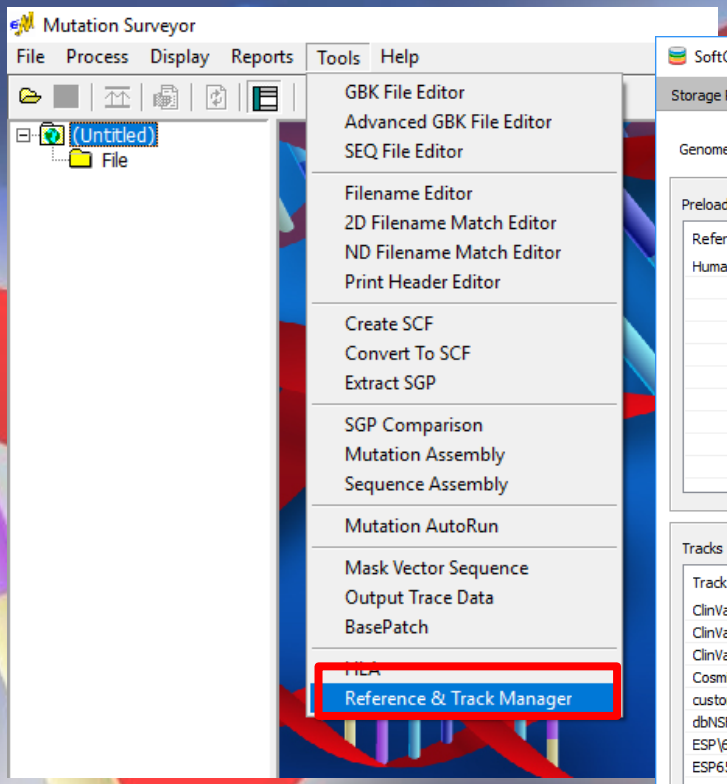


To get started, a whole human genome reference first needs to be imported. Please use the instructions on the following pages to complete this process. Projects will need to include GenBank files with chromosome coordinate information for display of Variation Tracks.

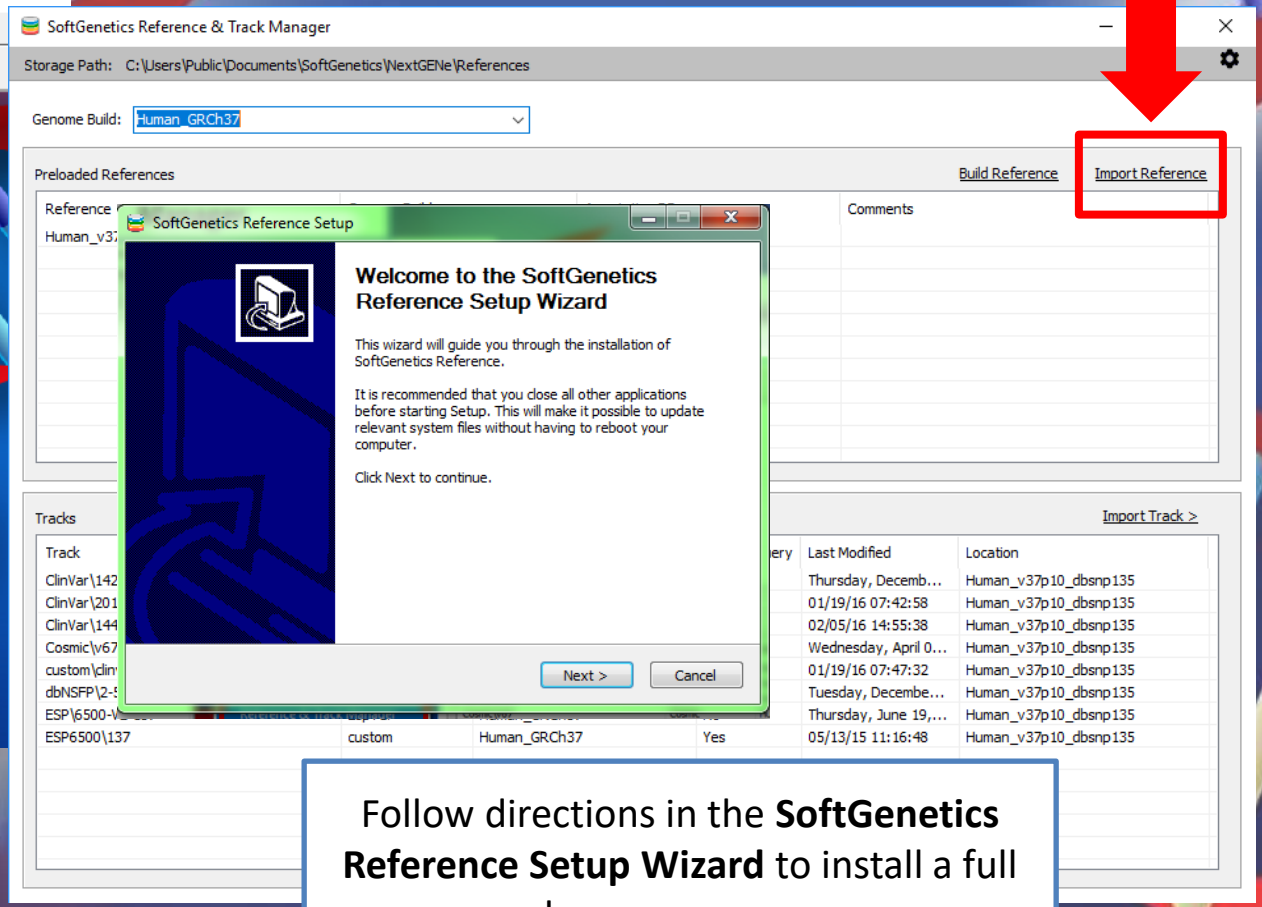
Note: Depending on the size of the files, the import process may require additional computer RAM and disk space and may take several hours to complete. Please contact us for more information at tech_support@softgenetics.com if interested in this feature.

Import Reference

In the main toolbar select **Tools → Reference & Track Manager**



Click on **Import Reference** link

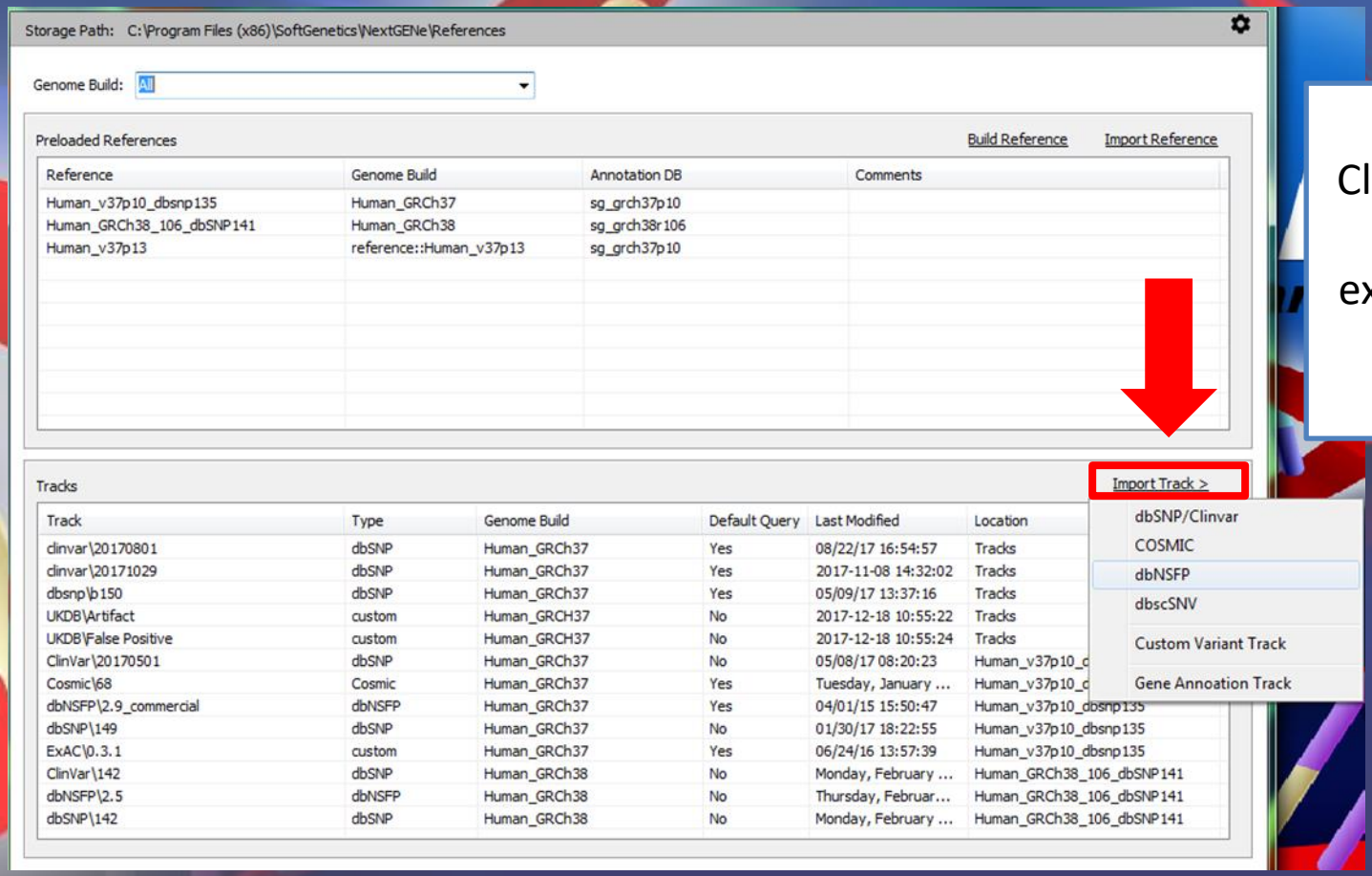


Follow directions in the **SoftGenetics Reference Setup Wizard** to install a full human genome

For detailed import guide please contact tech_support@softgenetics.com

Import External Database Annotation

In the main toolbar select **Tools → Reference & Track Manager**

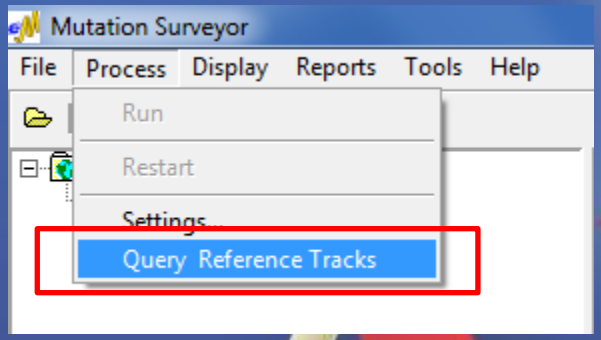


Click **Import Track** and select the external database to import

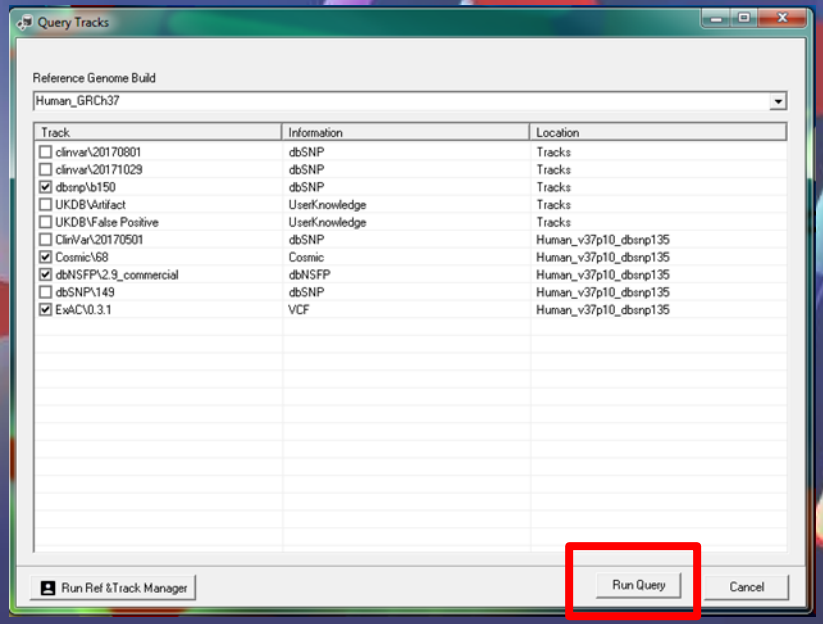
For detailed import guide please contact tech_support@softgenetics.com

Query Database Annotation

In the main toolbar select **Process→ Query Reference Tracks**



Select databases to query in Mutation Surveyor projects



For detailed import guide please contact tech_support@softgenetics.com