Quick Start Guide
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
Double click on files in the **Browser Pane** to view the trace data.
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**.
Graphic Analysis Display

Use icons to show/hide windows

Nucleotide, Amino Acid, and Database window

Trace windows

See page 13 for importing variation databases
By double clicking a mutation cell in the Mutation Report, the Graphic Analysis Display window appears.
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.
Heterozygous Indel Deconvolution Tool

Reference Trace
Sample Trace
Reference/Wild Type Allele
Mutant/Novel Allele
Mutant Allele Re-aligned to Reference
Location of Indel
Click the **Custom Report Builder** Icon in the main toolbar

Default settings group by Contig and pair Forward and Reverse files
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.
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