

Mutation Surveyor

Quick Start Guide



Opening Software for the First Time



Upload Data



Review Raw Data



Analysis Settings

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	File	Process	Display	Reports	Tools	Help
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Analyze Data



Graphic Analysis Display

Use icons to show/hide windows

 Mutation Surveyor

 File
 Process

 Display
 Reports

 Tools
 Help

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Graphic Analysis Display



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

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Heterozygous Indel Deconvolution Tool



Custom Report Builder

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Project Reviewer



Print Clinical Report

Mutation Surveyor	Click Clinical Banart Icon
File Process Display Reports Tools Help	in main toolbar
Adjust Display preferences Add a Custom Header file	Clinical Report Settings
Image: Weight with the second seco	Trace Display Range 20 bps
	3 ▼ Mutation Lais per Line 2 ▼ Lines per Page ✓ Show Reference Electropherogram □ Show Axes Values
Mutation Surveyor V5.1.1 2	Show Sample Electropherogram
Software: Nutation Storword Software: Nutation Storword Ave, Suite 500, State College: A 1600 USA Netsic Nutg://www.softgenetics.com Software: Nutg://www.softgenetics.com Software: Story: Softgenetics.com Software: Story: Software: Story: Softgenetics.com Software: Story: Softgenetics.com Software: Story: Softgenetics.com Software: Story: Software: Story: Story: Software: Story: Story: Software: Story: Story: Story: Story: Software: Story: Software: Story: Software: Story: Software: Story: Software: Story: Story: Story: Story: Software: Story: Story	/iew 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.

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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



For detailed import guide please contact tech_support@softgenetics.com

Import External Database Annotation

In the main toolbar select **Tools** \rightarrow **Reference & Track Manager**

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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

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