AMPLICON

Sequence Analysis

of Sanger Sequence and Ion Torrent PGM™ Sequencer Data



Next GENe®

Next Generation Sequencing Software for Biologists



Amplicon Sequence Analysis?

SoftGenetics is the obvious choice...

Mutation Surveyor® for Sanger Sequence Analysis.

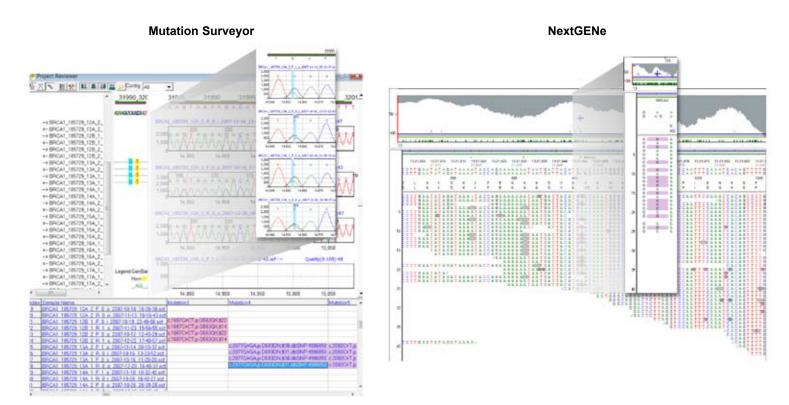
Patented "anti-correlation" technology provides 99.9% accuracy with sensitivity greater than 5% in the detection of SNPs, Indels and Mosaic variants from Sanger Sequencing traces generated by all major capillary electrophoresis platforms.

NextGENe®software for Ion Torrent PGM™ Sequence Analysis

Free standing Windows® based software for the rapid and accurate analysis of Ion Torrent PGM data. NextGENe utilizes point and click operation: requires no scripting, automatically sets analysis parameters for semiconductor sequencing data and provides results and analysis details in a highly graphical, user friendly interactive windows format.

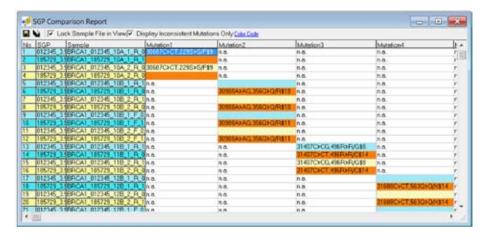
Softgenetics' Mutation Surveyor software's patented "anti-correlation" technology and simplicity of use has made it the leading DNA Variant Analysis software tool of Sanger sequencing reads worldwide. Our NextGENe software applies the same formula of unique technologies in an easy-to-use Windows® computing environment to the analysis of Next Generation Amplicon sequencing data.

NextGENe's proven technologies perform read quality assessment, automatically set analysis parameters based upon the data set and sequencing platform, perform alignment detecting SNPs and Indels with high accuracy and sensitivity, all in a biologist friendly, no scripting required interface.



Like Mutation Surveyor (left), NextGENe (right) analysis results are presented in a highly informative graphical and user friendly interface; logically presenting important biological information regarding the analysis and found variants.

Both NextGENe and Mutation Surveyor software programs include many tools to assist the biologist or clinician in interpreting, comparing, and researching results, all designed to enhance the analysis confidence in an efficient manner.



Mutation Surveyor's color-coded Comparison Report provides quick review of shared and negative variants between multiple patient sequences.



NextGENe's Rare Variant Analysis Tool compares familiy members or other groups indicated shared or negative mutations by patient, additional filtering by 1000 genomes frequency, and Polyphen score dramatically eases identification of rare variants.

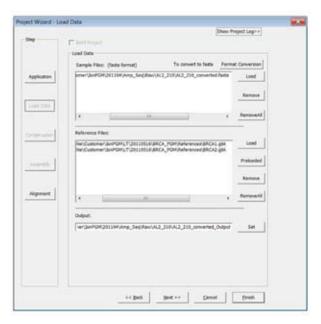
Biologist-Friendly Analysis Set-up

NextGENe and Mutation Surveyor were developed for use by the biologist and medical research clinician. Analysis set-up is simple and straightforward, no need for complicated scripting or informatics support. Simply follow the point and click Project Wizard to begin your analysis.

NextGENe contains unique technologies that examine your data, make necessary adjustments, and begin processing. Both programs have an automation feature which will process multiple projects sequentially. If you are using multiplexed Ion Torrent PGM data, the software automatically de-multiplexes and then will perform individual analyses on all of the sample sets.

Mutation Surveyor



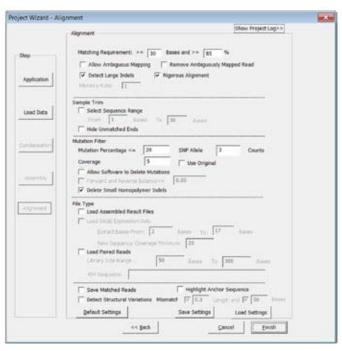


Adding Input data to be processed and selecting annotated references is simple and quick in both programs. Use the above dialogue box to select sequencing files, annotated reference and analysis output location. Both programs begin processing or further parameter optimizing can be accomplished within the advanced project wizard.

Mutation Surveyor Project Wizard

NextGENe Project Wizard

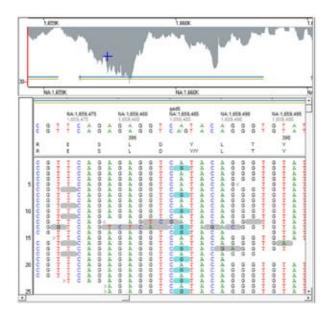


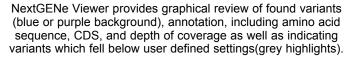


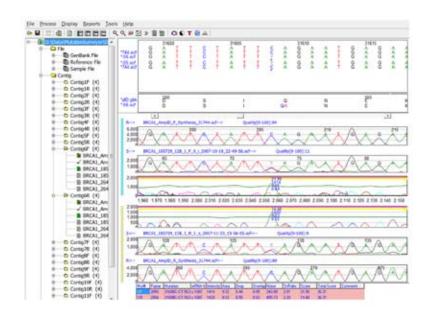
NextGENe's and Mutation Surveyor settings dialogue provide for complete control of the analysis. Advanced Users may wish to increase sensitivity, adjust to minimize false positives or, in the case of NextGENe, adjust the alignment stringency to detect large and small Indels. Default settings are applicable for most Amplicon data sets.

Usable/Informative Analysis Presentation

Analysis results in NextGENe and Mutation Surveyor are presented in highly graphical, succinct, interactive viewers. Graphical views are linked to customizable reports, which include gene, amino acid, links to dbSNP and other information such as depth of coverage in NextGENe, coding regions, mRNA, and regions of interest.







Mutation Surveyor presents found variants in the two central "mutation electropherograms" allowing immediate review of all found variations including SNPs, Indels and Mosaicism. Amino Acid Sequence, AA change as well as annotation are provided.

Comprehensive Reporting

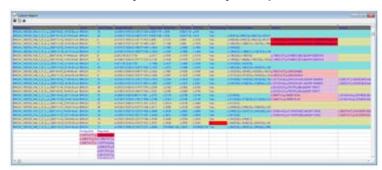
Analysis results are offered and reported with several filtering options to speed the review process. Edits to the analysis are permanently recorded in the project record. Each program offers specialized filtering options and are exportable in several formats for linking to LIMS systems or further investigation.

NextGENE's Comprehensive Mutation Report

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Included in NextGENe's reporting capabilities, all selectable by the user, are mutation confidence score, read balance, coverage, RNA Accession, mRNA, Amino Acid as well as Amino Acid changes, allele ratios and mutation calls. Previously reported variants and their dbSNP number are reported in purple for easy identification, novel findings are reported in blue. Previously reported variants include a direct link to the dbSNP database.

Mutation Surveyor Variant Analysis Report



Mutation Surveyor analysis reporting allows customizable reporting of variants by pathogenicity, reported or novel variants and other customer selectable formats.

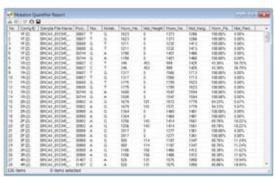
Unique Tools Enhance Analysis

NextGENe's Multiple Sample Comparison Tool



To facilitate discovery and/or clinical applications, NextGENe includes a comparison tool which allows up to 10 separate analyses to be compared to one another for similarities and differences. All views are linked for easy identification and review.

Mutation Surveyor's Low Frequency Quantification and Methylation Tool



Mutation Surveyor includes a unique quantification tool to measure percent change of mosiac, somatic and heteroplasmy variants and also methylation, based upon the intensity change in relationship to neighboring bases of the same wave length.

NextGENe's Expression Report

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NextGENe provides analysis information such as depth of coverage, similar to intensity value of Sanger Sequencing, normalized count values, as well as read directionality.

Acknowledgement

We wish to thank Halo Genomics AB for furnishing the Ion PGM BRCA data from their Selector BRCA Panel Kit.

For further information or to request a 30 day trial of NextGENe or Mutation Surveyor software please visit www.softgenetics.com or email info@softgenetics.com

Minimum Hardware Configuration:

PC: 32-bit Windows® XP OS, Dual Core Processor with 3GB RAM

MAC: Dual Core Processor, 32-bit Windows® XP OS, Boot Camp or VM Fusion, 3GB RAM

Optimal Hardware Configuration:

PC: 64-bit Windows® 7 OS, Quad Core Processor, 6GB RAM

MAC: Quad Core Processor, 64-bit Windows® 7 OS, Boot Camp or VM Fusion, 6GB RAM

SOFTGENETICS®

Software PowerTools for Genetics Analysis

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