SOFTGENETICS®

Program Portfolio

Support & Expertise

MUTATION Surveyor®
DNA Variant Analysis Software

NextGENe®
2nd Generation Sequence Analysis Software

Geneticist Assistant™
NGS Interpretative Workbench

GeneMarker®
The Biologist Friendly Software

GeneMarker® MTP
Multi-Template Processor

ChimeRMarker™
Automated Chimerism Analysis Software

GeneMarker® HID
Human Identity Software

Software ***PowerTools*** for Genetic Analysis

Sanger and Next Generation Sequence Analysis & Knowledge Base Development

MUTATION Surveyor®
DNA Variant Analysis Software

Mutation Surveyor® DNA variant analysis software for Sanger Sequencing is capable of performing variant analysis of up to 2000 Sanger sequencing files. Utilizing patented (US Patent 8,086,401) anti-correlation technology for detection of single nucleotide variations and performing automatic deconvolution of heterozygous insertions and deletions, Mutation Surveyor software delivers excellent accuracy, sensitivity, low false positive and negative rates in the analysis Sanger Sequencing reads.

Compatible with the output from all major capillary sequencing platforms, Mutation Surveyor software is unparalleled in the discovery of SNPS INDELS and Somatic Mutations.

Applications include:

- Direct Sequence Analysis
- Medical Sequence Analysis
- Mitochondria Sequence Analysis
- Resequencing Analysis

NextGENe®

2nd Generation Sequence Analysis Software

NextGENe® software is the perfect analytical partner for the analysis of desktop sequencing data generated by the ION PGM; Illumina MiSeq, Roche Junior as well as higher throughput systems as Illumina® Platforms and the Ion Proton™. NextGENe software provides a biologist-friendly scripting-free point & click interface, requiring little or no bioinformatics support. NextGENe software employs unique platform specific technologies in one free-standing multi-application package.

Application modules include:

- Variant Analysis (Targeted, WES/WGS) - SNP, INDELs, Structural variants, Somatic variant mining
- CNV-Seq
- RNA-Seq/Alternative splicing
- De novo assembly
- Expression - ChIP-Seq, miRNA
- Sensitive Aneuploidy Analysis
- HLA
- Forensic STR/mitochondrial analyses

SOFTGENETICS®

Software PowerTools for Genetic Analysis

Geneticist Assistant™

NGS Interpretative Workbench

Developed in collaboration with the Laboratory Medicine, Information Technology and Health Science departments of Mayo Clinic, **Geneticist Assistant** NGS Interpretative Workbench is a unique tool for the management, control, visualization, functional interpretation and historical knowledgebase development of NGS sequencing of Whole Exome data or Disease Panels targeted at specific genes for the purpose of identifying pathogenic variants associated with specific conditions such as hereditary colon cancers and others.

Geneticist Assistant Workbench is compatible with outputs from all major NGS sequencing platforms via standardized BAM and VCF file formats.

Information Included in single view:

- Functional Prediction information: SIFT, PolyPhen-2, LRT, MutationTaster, FATHMM, CADD & MutationAssessor
- Disease association: ClinVar & COSMIC
- Conservation scores: phyloP, GERP++, phastCons & SiPhy
- Population frequencies: 1000 Genomes and Exome Variant Server

Automatic Linkage to external databases, some requiring license:

- LOVD, Leiden Open Variant Database
- Alamut®
- HGMD®

Administration, real-time tracking of current project status, automated email notification and historical information.

Unique Tools:

- CAP Validation Assistance
- Automated BED file builder
- Positive Control Verification
- Point and Click Filtering
- Automated Comparison of Orthogonal Methodologies
- Informatics Pipeline

GeneMarker®

The Biologist Friendly Software

GeneMarker® is a unique genotyping software that has been designed to provide genetic researchers with a "biologist-friendly" genotyping tool. The program's linked-navigation and intuitive layout make it easy to use, while its accuracy, speed, and extensive collection of post-genotyping applications make it a powerful research tool.

GeneMarker software can perform analysis on up to 1,000 lanes of four or five color data sets generated by either slab gel or capillary electrophoresis. GeneMarker software is compatible with raw data files generated from custom chemistries kits and all major capillary and slab gel electrophoresis systems. GeneMarker software's embedded applications streamline analysis from raw data files through final reporting without any data transfer:

Basic Research Applications:

- AFLP®
- T-RFLP
- Microsatellite
- SnapShot® /SNPlex/SNPWave
- Clustering Analysis
- Kinship Analysis

Medical Research Applications:

- FragileX
- Trisomy/Aneuploidy
- MLPA®
- MS-MLPA®
- Cystic Fibrosis
- Microsatellite Instability

Programs for DNA Fragment & STR Analysis

GeneMarker® MTP

Multi-Template Processor

GeneMarker MTP software can apply up to six different analysis templates (size standard, panel, analysis parameters), allowing simultaneous analysis of CE plates with different chemistries. All of the GeneMarker software features are available for concurrent review, edits, saving/printing and post-genotyping applications.

Programs for DNA Fragment & STR Analysis

ChimeRMarker™

Automated Chimerism Analysis Software

ChimerMarker™ software, developed in collaboration with Dr. Donald Kristt, and validated by laboratories in the U.S. and Europe, combines accurate size and allele calls (genotyping) with automated Chimerism calculations and longitudinal monitoring. The program combines speed and accuracy with a biologist-friendly interface, typically saving up to 80% of analysis time.

ChimerMarker software features:

- Highly accurate size calibration and allele calling algorithms
- Ability to differentiate between donor, recipient, and shared peaks
- Automatic Calculation of Chimerism Percent
- Quality Metrics
- No Data Transfer required
- Long Term Monitoring of Chimerism level in allogeneic and autologous SCT, HSCT, BMT, post bone marrow engraftment, and PBSCT samples.

ChimerMarker software is compatible with all major STR chemistries and capillary sequencing platforms.

GeneMarker® HID

Human Identity Software

GeneMarker® HID Human Identity Software is an excellent choice for all human identity 4, 5 and 6 color STR analysis applications. This Expert System is designed with a user-friendly interface, audit trail, customizable access rights and tools that provide documented time savings (J Forensic Sci 56:29-35 2011).

Compatible with output from all Major Capillary Electrophoresis and Human Identification Chemistries.

Features:

Accurate sizing and allele calls to final reporting
Linked Applications:
Mixture Analysis
Paternity Analysis
Relationship Testing

Database Search Capabilities:

Crime Scene Investigation
Missing Person Identification
Mass Disaster Response
Paternity Testing

Rapid Response & Expertise

SoftGenetics knows if you have an analysis problem you want and need quick responsive technical assistance. Our promise is to assist each customer individually and rapidly. Over the years we have developed a reputation for the fastest and most comprehensive response to our nearly 2000 customers. If you need help, just email (*tech_support@softgenetics.com*) or call and one of our experts will reply in minutes.

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