SOFTGENETICS

Software *PowerTools* for Genetic Analysis

MUTATION Surveyor®
DNA Variant Analysis Software

NextGENe®
2nd Generation Sequence Analysis Software

GeneMarker®
The Biologist Friendly Software

GeneMarker® MTP
Multi-Template Processor

ChimeRMarker®
Automated Chimerism Analysis Software

Geneticist Assistant™
NGS Interpretative Workbench

GeneMarker® HID
Human Identity Software

GeneMarker® HTS
Forensic Analysis of Massively Parallel Sequencing

MaSTR™
Probabilistic Mixture Analysis of STR Profiles

Mitotyping Technologies

PROGRAM PORTFOLIO
Support & Expertise

**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 and 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
- Mitochondrial Sequence Analysis
- Resequencing Analysis
- Somatic Mutation Detection
- Methylation 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 S5 and PGM; Illumina MiSeq 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
- Batch CNV Analysis, a NGS replacement for MLPA®
- RNA-Seq/Alternative splicing
- *de novo assembly*
- Expression - ChIP-Seq, miRNA
- HLA
- Trio or Family Comparison

SOFTGENETICS®
Software PowerTools for Genetic Analysis

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 to six color data sets generated by either slab gel or capillary electrophoresis. GeneMarker software is compatible with raw data files generated from commercial and custom chemistries and commercially available sequencing platforms.

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 /Dendograms
Kinship Analysis /Database search

Medical Research Applications:
Repeat Expansion, such as HTT & DMPK
FragileX
Trisomy/Aneuploidy
MLPA®
MS-MLPA®
Microsatellite Instability
Cystic Fibrosis

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.

ChimeR Marker ®
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 includes a Maternal Cell Contamination (MCC) detection and quantification module

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

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 gnomAD

Automatic Linkage to external databases, some requiring license:

LOVD, Leiden Open Variant Database
Alamut®

Unique Tools:

Artifact Tracking
CAP Validation Assistance
Copy Number Variation (CNV) support
Automated BED file builder
Positive Control Verification
Point and Click Filtering
Automated Comparison of Orthogonal Methodologies
Informatics Pipeline
Customized Reporting

GeneMarker® HID
Human Identity Software

- NDIS Approved Expert System
- 4 - 6 Dye Capability
- Fast, Accurate, and User-friendly
  - Documented time savings of up to 40%
  - Up to 70% less analyst intervention
- High Capacity
- User Management with Audit Trail
- Validation Assistance - Automated Analytical Threshold Determination
  - Linked Applications
  - CODIS Reporting
  - Mixture Analysis – two-person deconvolution and likelihood ratios
- Relationship Testing /Kinship
  - Paternity (per AABB Standards)
  - Database Search Tools and Likelihood Ratio Results:
    - Missing Persons Search
    - Mass Disasters
    - State DNA Index System (SDIS) Search
  - Contamination check – percent same genotype:
    - Autosomal & Y-STR Searches
    - Sample – to – Sample in same project
    - Sample – to – database comparison
- Compatible with all Major CE and Rapid Systems
- Windows ® 7 – 10 compatibility

GeneMarker®HTS
High Throughput Sequencing Software
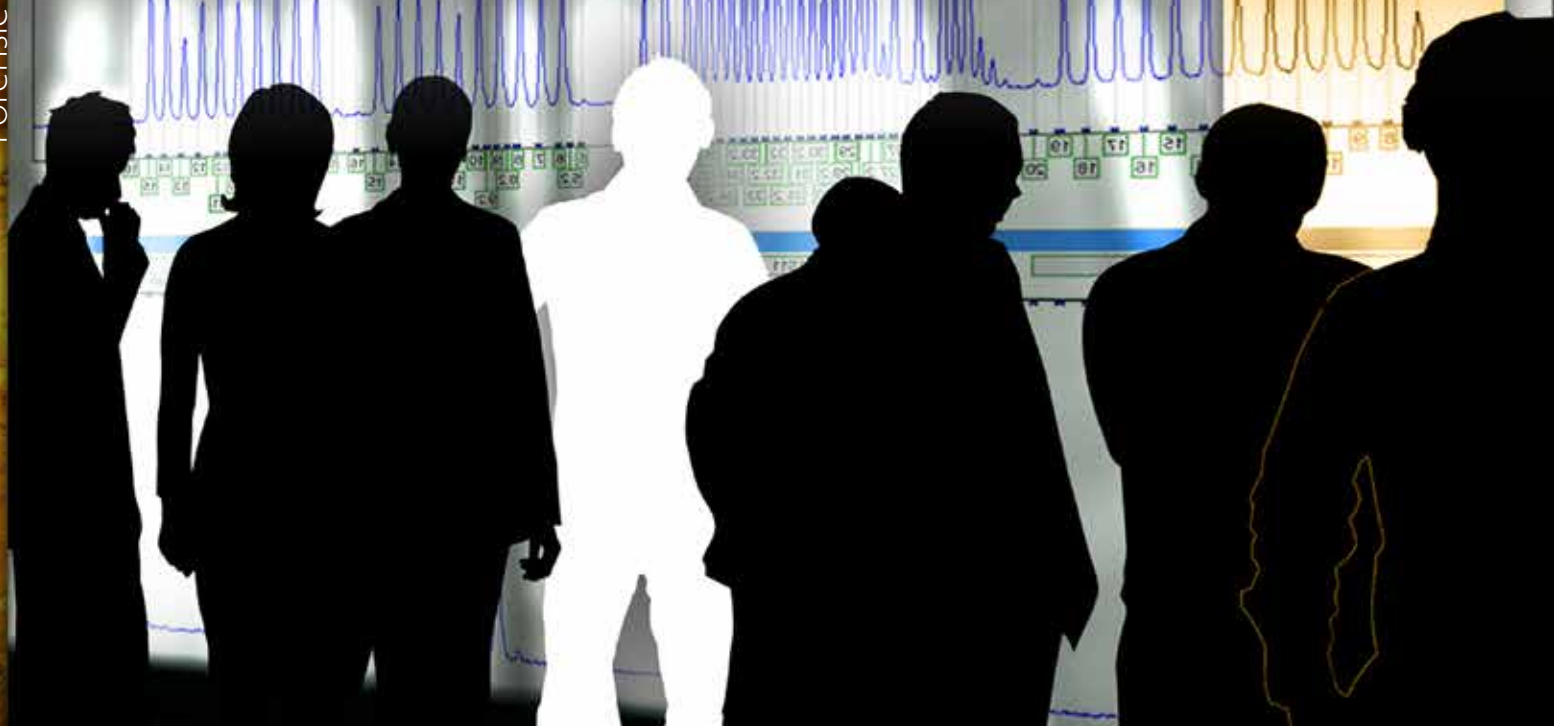
### *Analyze mtDNA & STR/Y-STR's Simultaneously from MPS reads*

Validated Software in an Easy-To-Use Windows Interface which is compatible with Major Chemistries and Platforms. Includes Comprehensive Reporting Options, as well as full Audit Trail and Administrative /User control.

STR Analysis Features:

• Autosomal & Y-STRs

• Forensic Nomenclature

• Genotype & Iso-Allele Reporting

• Meets the SWGDAM Interpretation Guidelines for Autosomal STR Typing by Forensic Laboratories to Address Next Generation Sequencing

• **Simultaneous mtDNA Analysis Features:**

• Whole Genome or HV1/HV2 & Control Region Analysis

• Major and Minor Variant Reporting (SNPs, Indels)

• Unique mtDNA Alignment Technologies:

      -Motif

      -Consensus

• Forensic Nomenclature

• Easily Uploads to EMPOP

MaSTR™

Probabilistic Mixture Analysis of STR Profiles
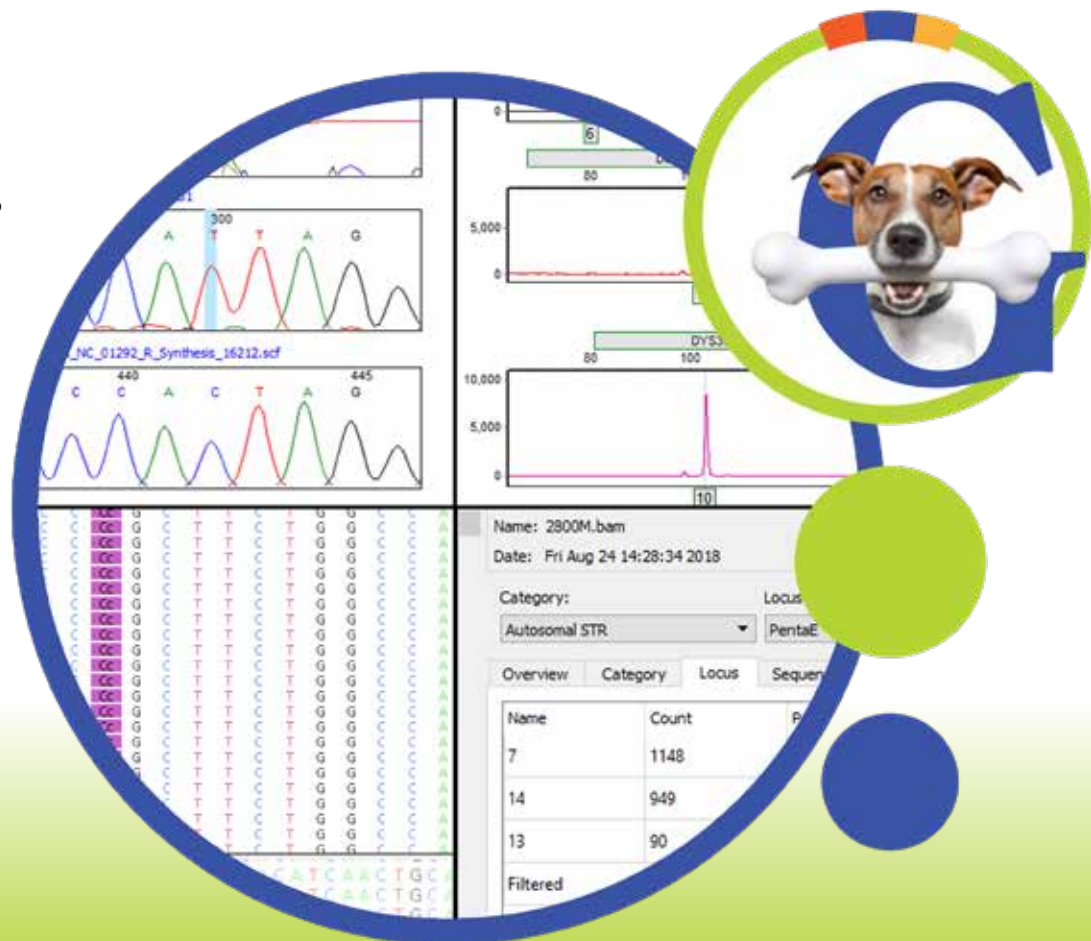
MaSTR™ Enlightened Probabilistic Mixture Analysis of STR Profiles software

MaSTR software features a rapid and transparent approach to Probabilistic Mixture Analysis which utilizes your forensic acumen in an easy-to-use Windows® environment for research, validation and casework applications. Server based MaSTR software is very cost effective, includes two simultaneous user licenses, and automatic queuing of multiple analyses.

MaSTR Software features:

- Transparency, Complete Documentation including Analysis Code to users
MaSTR software was designed following *The Organization of Scientific Area Committees for Forensic Science (OSAC) and Scientific Working Group on DNA Analysis Methods (SWGDAM)* probabilistic guidelines and published probabilistic approaches (Taylor, *et al.* and Bright *et al.*). The analysis code is available, on a confidential basis, with purchase of the software.

- Validated
Contact info@softgenetics.com for a copy of the validation study performed by Dr. Michael Adamowicz. Dr. Adamowicz is currently Director of the Forensic Science program at the University of Nebraska and was previously a forensic science program coordinator at the University of New Haven, Henry C. Lee College of Criminal Justice & Forensic Sciences and a member of the SWGDAM Mixture Committee developing / writing mixture analysis guidelines.

- Lower Acquisition & Operational Costs
Single server-based program has an exceptional capacity to meet most forensic laboratory needs. The initial license includes two concurrent users. Low cost, additional clients allows each analyst station to have access to MaSTR software.

- Administration Tools and Modes of Operation
MaSTR software requires the user to log in to access the software functions. This requirement allows the laboratory to password protect their individual models and mixture analysis results.

- Flexible Capacity
Unique System Design -- MaSTR probabilistic genotyping software makes efficient use of the lab computer resources. With Server-client based technology, processing is done on the server and multiple clients can review results and send new jobs. Each user sets up and sends analysis jobs from *existing* client computers. Expedited cases can be moved to the front of the queue by laboratory management.

- Technology
MaSTR software's unique, easy-to-use interface enables analysts to create and submit mixture analysis jobs in minutes. The server-client configuration provides flexibility and lowers costs by performing the analyses on a dedicated computer, freeing up the analyst's time and computer for other tasks.

- Customizable Reporting
Each report contains a cover page with the institution name and logo, a table of contents and the fields selected by the analyst.

Enlightened Probabilistic Mixture Analysis Software

Mitotyping Technologies

Mitotyping Technologies, the clear choice in forensic mitochondrial analysis

We are Single-minded: Mitotyping Technologies is one of the oldest and most experienced group of scientists devoted to this forensic and anthropological DNA specialty. We have worked on criminal, civil, exoneration and federal cases in all 50 states and numerous foreign countries. We have testified over 200 times in mtDNA cases for both the prosecution and defense teams.

Ancient DNA Methodology for Highly Degraded evidence:

Mitochondrial DNA Analysis of shed hairs and hair fragments is our specialty. We have a >95% success rate developing profiles from hairs of all ages and sizes. For highly degraded samples, we offer an "ancient DNA" approach. Each sample is analyzed individually to account for its specific analytical needs.

Considerations in selecting a mtDNA Laboratory:

Is Lab fully accredited for mtDNA analysis?
(We have been since 2001)

What is their success rate?
(Ours is >95% for hair evidence, 90% overall)

How much experience do they have?
(Our Senior Scientist has over 19 years)

Do they batch or process evidence individually?
(Each sample is processed individually, we never batch process)

Is expedited service available?
(We will provide expedited services with the same precision and accuracy as provided with normal casework. Please contact us for more information.)

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.
Commercial inquiries: info@softgenetics.com

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