SOFTGENETICS®
Software *PowerTools* for Forensic Analysis

GeneMarker®HTS

Forensic NGS Analysis Software

Analyze mtDNA and STR/Y-STRs Simultaneously

Now Includes:

Mitochondrial DNA Analysis

- ✓ Whole Genome or HV1/HV2 and Control Region Analysis
- ✓ Unique Alignment Technology[1,2] • **Motif • Consensus**
- ✓ Forensic Nomenclature
- ✓ Easy uploads to EMPOP[3]

STR Analysis

- ✓ Autosomal & Y-STR
- ✓ Forensic Nomenclature
- ✓ Genotype & SNP Reporting

Validated

Easy-to-Use Windows® Interface

Compatible with major Chemistries & Platforms

Audit Trail & User Control

Comprehensive Reporting Options

SOFTGENETICS®
Software *PowerTools* for Forensic Analysis

GeneMarker® HTS

Forensic NGS Analysis Software

Mitochondrial DNA Analysis

GeneMarkerHTS software provides a validated, streamlined workflow for forensic casework and medical research of mitochondrial DNA data analysis from massively parallel sequencing (MPS) systems such as the Illumina® and Ion Torrent® platforms; in an easy to use Windows® operating system with password protected user rights and administrative controls/ audit trail. Developed in collaboration with leading laboratories, GeneMarkerHTS software provides rapid analysis of multiple samples using consensus alignment or a unique motif alignment technology that automates the recommendations of ***DNA Commission of the International Society for Forensic Genetics: Revised and extended guidelines for mitochondrial DNA typing***. Using forensic motif alignment provides recognition and proper assignment of motifs and INDELs consistent with phylogenetic and forensic considerations.

Analysis results include:

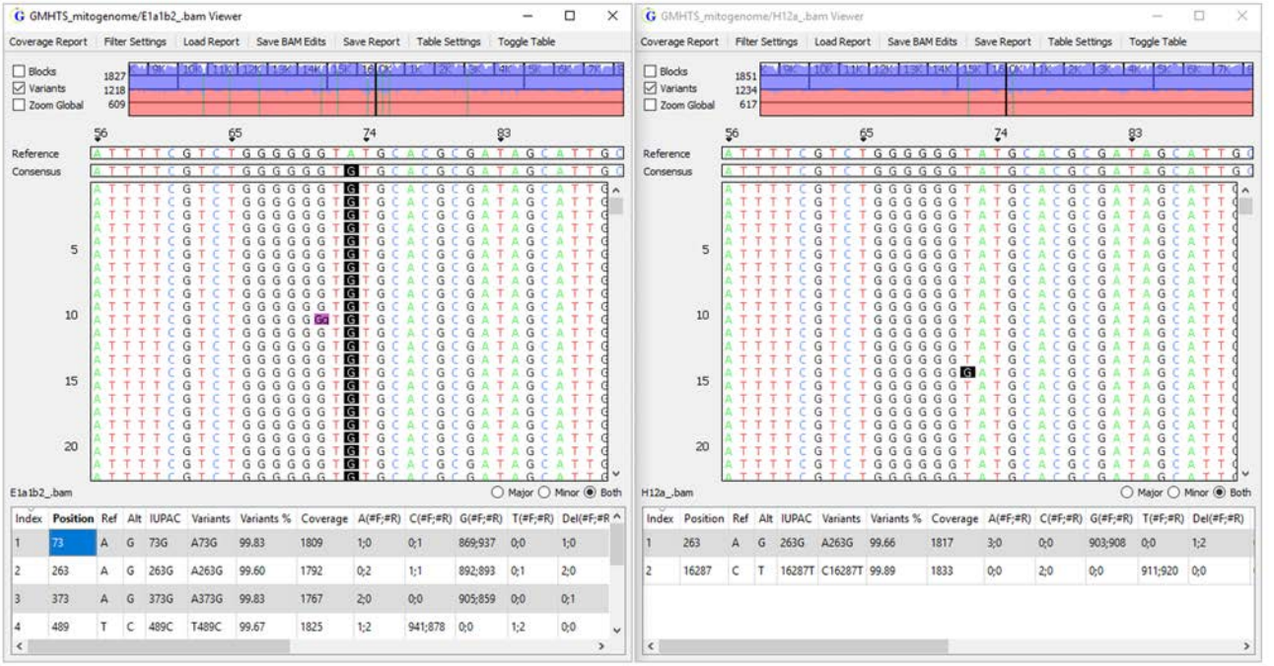
- Consensus sequence, Variants, SNPs, Indels
- Depth of coverage graphics
- Major variant report for haplogroup determination, Import to EMPOP[3] (ENDAP mtDNA Population Database)
- Consensus sequence aligned to reference (IUPAC nomenclature)
  - o Whole mtDNA genome, spanning the origin
  - o Specified areas of interest, such as control region, HV1, HV2
- Read pile-up (with depth and direction indicators)
- Compare multiple samples in single view
  - o Synchronized view, scroll and zoom of multiple samples
  - o Comparison viewer, table with sample-to-sample and and variant composition

Global and zoom example of a whole mtDNA genome alignment:

Global (left) and zoom example (right) of a whole mtDNA genome alignment: The Global View shows the depth of coverage; forward read coverage in blue and reverse read coverage in red, Reference sequence and Consensus Sequence are above the pile-up of the reads, linked result table is located under the read pileup. The large black rectangle in the pileup (left) is the magnified region (right).

Rapid Analysis and Synchronized Viewing of Multiple Samples

GeneMarkerHTS software provides results in minutes; for example, 30 MiSeq whole mtDNA chromosome data files with 10,000 average depth of coverage were aligned in 90 minutes (3 minutes per sample). In a more extreme example, 200 whole mtDNA chromosome data files with 10,000 average depth of coverage aligned in 16 hours.

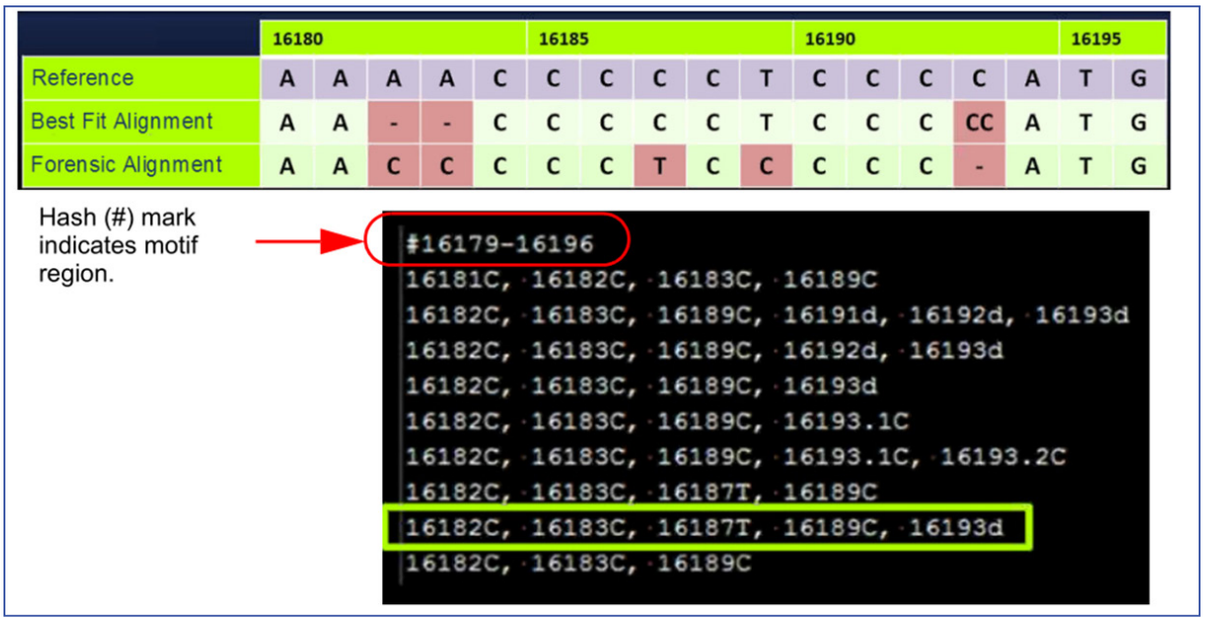


Example of synchronized viewing of two samples, E1a and H12a: all open samples are synchronized when the analyst clicks on a position or variant in the table or zooms/scrolls in the pile-up. Up to four samples can be easily reviewed on most monitors, with larger monitors accommodating up to 8 open samples for synchronized viewing and review.

Time saving tools

Motif alignment reduces manual edits for forensic alignmemt[1,2]

GeneMarkerHTS software has an extensive, preloaded forensic motif file as well as a motif editor to assist labs in adding new motifs. Motif alignment technology automates the recommendations of the DNA Commission of the International Society for Forensic Genetics and decreases analyst intervention.



Comparison Viewer and Reporting:

Compare results between samples of a project after alignment, or samples selected from previous projects in the database.

Variety of reports:

- Consensus sequence
- Variant reports – SNPs, insertions and deletions
- Haplotype, heteroplasmy
- Report compatible for import into EMPOP[3]

The software includes:

- Audit trail capability
- User management
- Customizable viewing and reporting to protect privacy of potential health information (PHI) sequences
- Comparison Capabilities

Comparison Viewer is a viewing tool to compare analysis results of multiple samples. Use the comparison viewer for:

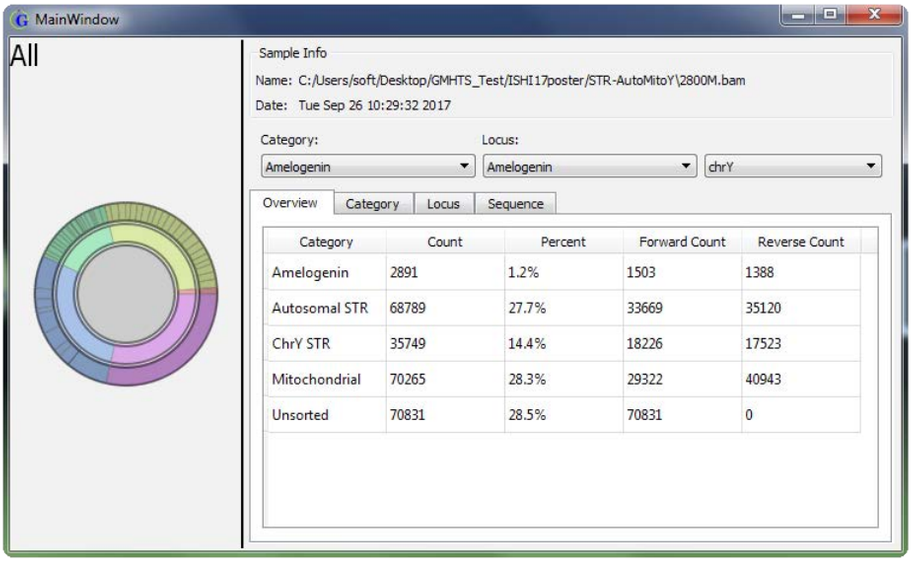
1. Sample to sample comparison (above)
2. As well as a variant comparison of all the samples in the project at the same time (below)

Autosomal and Y-STR Genotypes and SNPs

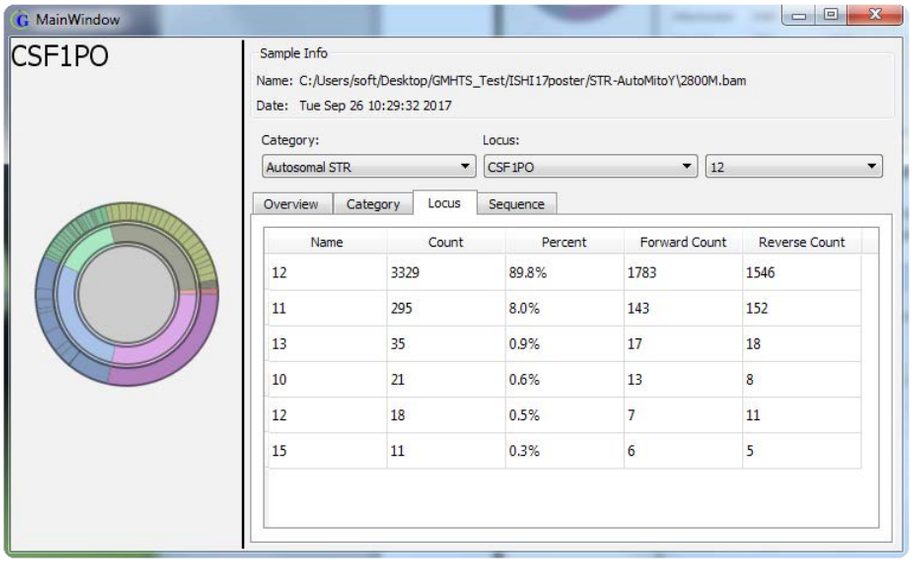
High-throughput sequencing data for forensic applications, database or casework, can be analyzed by selecting a built-in panel or by loading a panel for custom chemistries. Autosomal and Y-STR analysis includes conventional forensic nomenclature. **Contact info@softgenetics.com for a copy of the concordance study.** GeneMarkerHTS software capabilities include simultaneous analysis and reporting of mtDNA and STR chemistries.

Review and save reports at different levels of details **-- no need to scroll through pages of results to locate the area(s) of interest.**

- Overview – Assignment of reads to different analysis categories (Autosomal, CHR Y, …)
- Category – Assignment of reads among the loci in category
- Locus- frequency and identified names of sequences in the selected locus
- Sequence – detailed quantitative information for the selected sequence along with annotation information



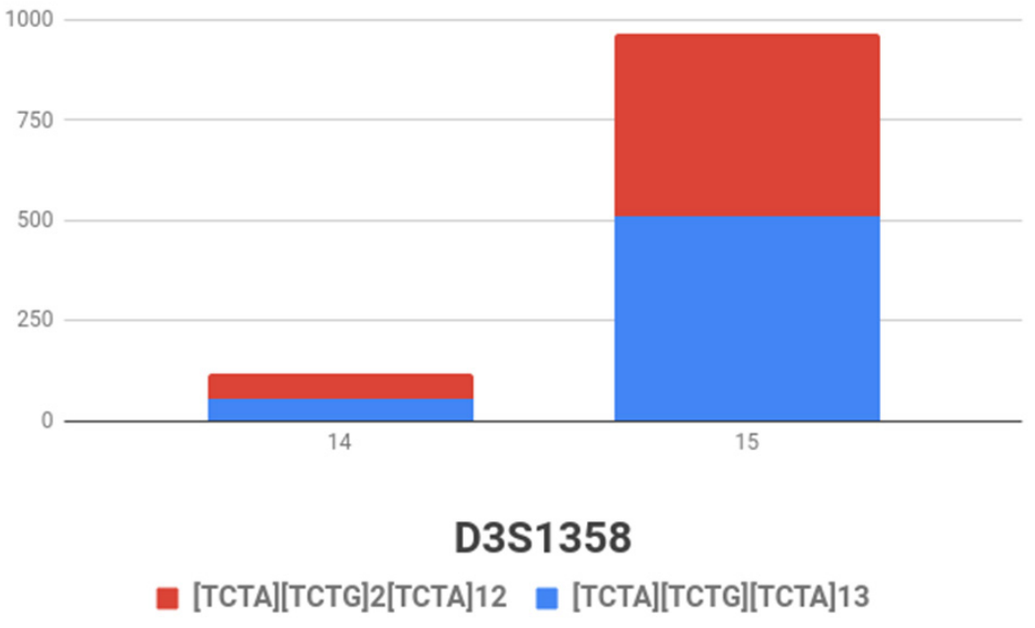
The Overview results are initially displayed allowing a comprehensive review of the results to start. The results can be broken down by selecting a category and locus from the dropdown lists.



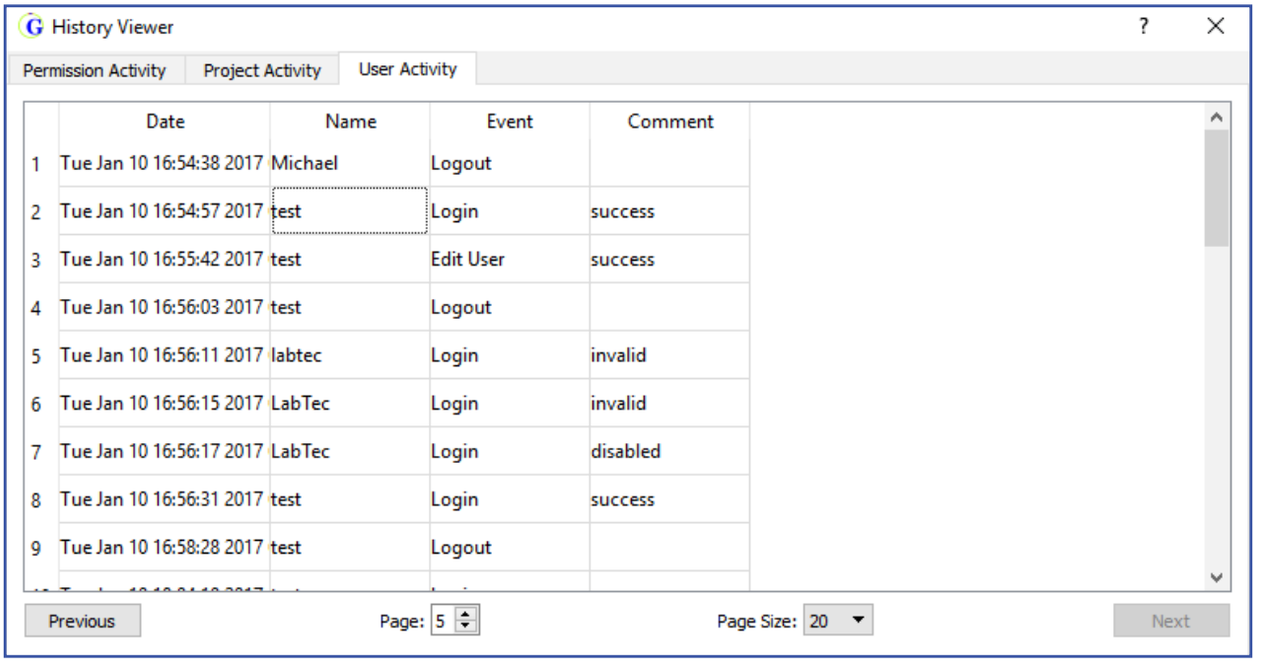
The Locus tab displays results for each allele in the selected locus.

Visualization of Isoalleles

Isoallele: A locus that appears homozygous in length-based measurements (such as CE), but is heterozygous by sequence. High throughput sequencing provides allele calls and sequence variants. ***This depth of information has applications in identification of individuals and relatives in single source samples and the potential for improved assignment of alleles to contributors during analysis of mixtures.***



User management and control of access rights is managed by the laboratory administrator



The database provides password protected access rights, audit trail and allows users to upload/download projects with the initial analysis parameters and upload/download changes to analysis parameters of subsequent analysts. The database provides a record of all analysis parameters and activities on a data set.

Minimum Recommended processing hardware:

64 bit Windows OS
12 GB RAM
2.4GHz Dual Quad Core Processor

If trial disc is not present please email info@softgnetics.com
for a free 30-day trial

[1] Parsons et al. 2014 Forensic Science International: Genetics. 13:134-42

[2] Holland, Pack and McElhoe. 2017 Forensic Science International: Genetics. 28:90-98

[3] European DNA Profiling Group (EDNAP) ENDAP mtDNA Population Database https://empop.online/

Thank you to Promega Corporation, Madison, WI, USA for providing Autosomal and Y-STR data.

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