

CONCORDANCE STUDY: HIGH-THROUGHPUT STR/YSTR RESULTS USING GENEMARKER®HTS SOFTWARE ARE CONCORDANT WITH CAPILLARY ELECTROPHORESIS RESULTS AND PROVIDE ADDITIONAL SEQUENCE VARIATION INFORMATION

K. Hendricks, C.S. Liu, L. Luo, J. McGuigan, T. Snyder-Leiby, N. Wan, J. Wu
SoftGenetics, LLC

GeneMarker®HTS software was released in 2016 as a rapid, user-friendly, software for forensic mtDNA analysis. Several Forensic Science International: Genetics articles evaluating and using the software were published in 2017 (Holland et al, Calloway et al, Riman). Development of autosomal and Y-STR capabilities started in 2017; providing genotype and single nucleotide polymorphism (SNP) detection within amplicons.

While mtDNA analysis is essential in many forensic applications, STR DNA analysis impacts countless investigations and court cases. Strengths of this data include both its resolving power for excluding an individual and the ability to determine potential relationships between evidence and suspects due to Mendelian inheritance of nuclear DNA. High throughput sequencing methods provide additional resolving capabilities in situations where two individuals have the same allele sizes with differences in nucleotide repeat sequence.

To ensure continuity with the well-established capillary electrophoresis (CE) nomenclature, it is essential to confirm concordance of MPS analysis results with CE results. This presentation details a concordance study between GeneMarkerHTS STR/YSTR results and the corresponding samples analyzed by capillary electrophoresis.

National Institute of Standards and Technology (NIST), in conjunction with Promega corporation, generously supplied the fastq sequence files and the corresponding CE allele calls for 672 samples amplified with the PowerSeq® Auto/Y System and analyzed on an Illumina® MiSeq. Results of these data analyzed in GeneMarkerHTS software were highly concordant with the CE allele calls. Summary of the allele calls concordance and examples of instances where alleles exhibited sequence variation will be presented.

Chemistries for mtDNA and STR amplification for HTS platforms enable the laboratory to have the benefits of both mtDNA and STR analysis at the same time. In addition to identification of sequence polymorphisms, advantages of HTS STR chemistries over traditional CE STR chemistries include the ability to have smaller amplicons and to analyze more loci in one reaction. GeneMarkerHTS is rigorous, user-friendly software for the analysis of HTS data for STR and mtDNA applications.