GENEMARKER®HTS (HIGH THROUGHPUT SEQUENCING) SOFTWARE UPDATE – NOW INCLUDES SHORT TANDEM REPEAT (STR) ANALYSIS CAPABILITIES

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GeneMarker®HTS software was introduced 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). **The new release of GeneMarkerHTS software includes rapid analysis and reporting for HTS STR data** in addition to the mtDNA analysis capabilities.

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, such as Illumina® and Ion Torrent™, provide additional resolving capabilities in situations where two individuals have the same alleles, but slight differences in nucleotide repeat sequence. 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 analysis of HTS STR and mtDNA data.**

GeneMarker®HTS software provides a streamlined workflow for forensic mitochondrial DNA data analysis from High Throughput Sequencing (HTS)/ Next Generation Sequencing(NGS) systems such as the Illumina® and Ion Torrent™ platforms; in an easy to use Windows® operating system. 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 fulfills recommendations for forensic alignment, and provides recognition and proper assignment of motifs and INDELs consistent with phylogenetic and forensic considerations.

Workflow and data analysis of sequences generated with Promega PowerSeq™ system and Applied Biosystems™ *Precision ID* chemistries; Illumina® MiSeq and ThermoFisher Ion S5 platforms will be presented along with a review of the mtDNA genome forensic alignment, heteroplasmy report, import of major variant report to EMPOP, sample comparison reporting and database options.