CONVERGE™ SOFTWARE – A CONCORDANCE STUDY COMPARING CE-BASED SYSTEMS AND NGS TECHNOLOGY USING STR LOCI WITH STANDARD DATA SETS

S Vijaychander [1], A Swami [1], S Mehta[1], P Dakle[1], M Gundapuneni [1], N Rajagopalan [1], M Karpagavinayagam [1], J Deng [1], R Gupta [1]
[1]Thermo Fisher Scientific

CE-based forensic DNA workflows are the mainstay of DNA typing, although limitations exists in terms of low capacity of multiplexing, difficulty in mixture interpretation, partial or no profiles from highly degraded samples and laborious mtDNA sequencing. With the introduction of Next Generation Sequencing (NGS) technology, its workflow allows more genetic information, multiplex many types of markers simultaneously and target amplicons even smaller than those found in CE mini-STR kits across challenging samples. Due to complexity of the two platforms, there is a vital need to cross-functionally manage and validate data.

Converge™ is a comprehensive software platform that integrates CE and NGS workflows and compares genotype calls from disparate systems; unifying the operational view to provide confidence in the results. A series of single source and mixture experiments were performed to evaluate genotype accuracy, depth of coverage and allele balance as additional informative metrics for assessing quality of data.

The data set comprised of 37 samples including 12 single source and 25 mixture samples with varied mixture proportions of eight contributors using an input DNA of 0.025ng for minor to 1ng of major contributor. Early Access (EA) GlobalFiler™ NGS Mixture ID panel was designed to detect 33-plex STR loci, including expanded Combined DNA Index System (CODIS) core loci (CSF1PO, D1S1656, D2S1338, D2S441, D3S1358, , D5S818, D7S820, D8S1179, D10S1248, D12S391, D13S317, D16S539, D18S51, D19S433, D21S11, D22S1045 TH01, TPOX, FGA , vWA, DYS391, AMEL-X and AMEL-Y) and 9 non-CODIS loci (D1S1677, D2S1776, D3S4529, D4S2408, D5S2800, D6S1043, D6S474, D12ATA63, and D14S1434), in addition to identity SNPs and micro haplotypes, with NGS technology on the Ion-S5™ system.

The results from samples processed on the S5 system were compared to samples analyzed with GlobalFiler Express kit and GeneMapper® *ID-X* v1.5 software. For single source samples, excluding SE33 from CE analysis, 14/22 common STR makers showed 100% accuracy and remaining 9 markers showed >96% accuracy. Any deviations observed could be attributed to various aspects including, but not limited to, optimizing thresholds with NGS data or SNPs in flanking region.

The promising nature of the results from this study not only demonstrates the advances in NGS technology for forensics but also huge utility of Converge software platform that enables a streamlined workflow with fast turn-around time.

For Forensic or Paternity Use Only