EVALUATION OF THE ION PGM™ SYSTEM FOR USE IN HUMAN IDENTITY DNA TYPING

Churchill JD[1], Chang J[2], Ge J[2], Rajagopalan N[2], Lagacé R[2], Liao W[2], King JL[1], Budowle, B[1,3]

[1]Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center
[2] Human Identification, Thermo Fisher Scientific
[3]Center of Excellence in Genomic Medicine Research (CEGMR), King Abdulaziz University

The introduction and continued improvement of massively parallel sequencing (MPS), also referred to as next generation sequencing, technologies offer the scientific community novel and enhanced approaches to DNA typing. While capillary electrophoresis-based technologies have been the standard method for human identity typing applications, there are limitations in this methodology's resolution, scalability, and throughput. MPS has the potential to address these limitations and expand forensic typing capabilities. MPS provides the ability to multiplex multiple types of forensically relevant genetic markers and sequence multiple samples per run. Additionally, MPS data may improve interpretation of mixtures. The Ion Torrent Personal Genome Machine ® (PGM™) (manufactured by Life Technologies, a Thermo Fisher Scientific Brand) is one available MPS benchtop platform. The PGM is a high-throughput sequencer that employs semiconductor sequencing technology with customized chemistries in a laboratory workflow that enables high-throughput and fast run times at a reasonable cost.

Twelve blind genomic samples, containing total DNA ranging from ~42 to 280 ng, were provided by a third party (the Green Mountain Conference) for genetic analysis. Three separate Ion PGM™ panels containing human identity SNPs, ancestry informative SNPs, and STRs, as well as the whole mitochondrial genome, were run and analyzed for these 12 samples. All four genetic systems were run and analyzed on the Ion PGM™ system in a reasonably quick time frame. Completeness of genetic profiles, depth of coverage, strand balance, and allele balance were evaluated as informative metrics for the quality and reliability of the data produced. All results from analysis of the 12 genomic samples were consistent with sample information provided by Green Mountain at the end of the blinded study. The promising nature of these results warrant full validation studies of these MPS technologies and continued development of tools for data analysis.