APPLICATIONS OF ION TORRENT PGM™ IN HUMAN IDENTIFICATION

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Next-generation sequencing technology is a subject area in which the forensic community has previously done little research. However, this presentation shows that using genomic DNA or mitochondrial DNA extracted from bone, blood, buccal swab or other forensic samples, one can obtain mitochondrial, SNP and/or STR genotypes multiplexed in a single next-generation sequencing run.

To address next generation sequencing needs of the forensic community we have developed a series of applications for the PGM™ Sequencer.

For mitochondrial DNA we have two protocols, one for the complete genome, and another using small amplicons spanning the hypervariable region. For instances where DNA is degraded or there are privacy issues the mini amplicon set may be more appropriate. To demonstrate feasibility both systems were tested against reference samples and the results reported.

Two SNP panels have been designed, one focused on human identity and leveraging SNPs from the public SNP sets of Ken Kidd and SNPforID, and another focusing on investigative leads comprised of phenotypic (hair, eye color, skin color) and ancestral SNPs. Performance enhancements for each panel will be discussed in relation to the new sequencing protocols, chemistries and chips, now available for the PGM™.

A multiplex STR panel for concurrent use with SNPs has been developed to enable linking to existing STR databases along with sample tracking in addition to the SNP data that can be obtained with the SNP panel alone. Concordance results with current CE STR technologies will be evaluated.