INCREASING DISCRIMINATION OF DEGRADED DNA USING QUANTIFILER® TRIO WITH THE ION PERSONAL GENOME MACHINE® SEQUENCER

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Forensic casework analysis that involve degraded or compromised DNA samples in trace amounts often show allele dropouts of the larger molecular weight loci, which result in partial or no probative results. It is beneficial to have accurate DNA quantification data to enable forensic analysts to critically evaluate how to select the appropriate downstream technologies in order to obtain the most probative results with challenging casework samples. At the threshold that STR kits cannot provide complete profiles anymore, forensically informative SNPs can provide the additional discriminatory power to complement partial or no STR profiles produced by capillary electrophoresis (CE). The HID-Ion AmpliSeq™ Panels were developed to provide an alternative and complementary approach to current human identification STR technologies, as well as utilize Next Generation Sequencing (NGS) high-multiplexing capabilities.

The HID-Ion AmpliSeq™ Identity Panel consists of 90 autosomal and 34 upper Y-clade markers. These SNPs were selected due to their high heterozygosity and low population heterogeneity. Using an input of pristine DNA, each independent SNP provides a random match probability of 10^{-33} in most parts of the world. The 34 Upper Y-Clades were selected from the Y-Phylogenetic Tree[1].

The HID-Ion AmpliSeq™ Ancestry Panel consists of 55 Kidd[2] and 123 Seldin[3] markers. These SNPs were selected to infer ancestry from the major eight global regions. These SNPs are highly supported by Kidd's ALFRED[4] database, which contains a considerable amount of allele frequency variation and population statistics.

We set out to evaluate data to determine how to proceed with sample processing for degraded DNA samples. Mock DNA of varying degrees of degradation was produced using both mechanical and enzymatic shearing techniques. Then, utilizing the Degradation Indices (DI) provided by Quantifiler® Trio DNA Quantification Kit, a plot of Probability of Identity (PI) vs DI was drawn to illustrate the best downstream sample processing for samples identified with varying level of degradation. We showed a correlation between the calculated DNA degradation level and probative value obtained from the PGM™ SNP results. We further demonstrated that the HID SNP panels correlate to CE STR data but offer a higher discrimination power to make identifications from samples that would be considered "poor" quality for STR processing in a casework laboratory.

1 Karafet et al. New binary polymorphisms reshape and increase resolution of the human Y chromosomal haplogroup tree. Genome Res (2008) vol. 18 (5) pp. 830-8

2 Kidd, K. K. (2012). Better SNPs for Better Forensics: Ancestry, Phenotype, and Family Identification. Poster

3 Kosoy, R., Nassir, R., Tian, C., White, P. A., Butler, L. M., Silva, G., et al. (2009). Ancestry informative marker sets for determining continental origin and admixture proportions in common populations in America. Human Mutation, 30(1), 69–78