DNA FORENSIC MARKER ANALYSIS WITH THE ABBOTT PLEX-ID SYSTEM

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The Abbott PLEX-ID system provides a fully-automated platform for electrospray ionization time-of-flight mass spectrometry analysis of PCR-amplified nucleic acids (PCR/ESI-MS). Assays for mitochondrial DNA (mtDNA) profiling, short tandem repeat (STR and Y-STR) analysis, and human autosomal SNP analysis have been developed that can be analyzed automatically on the same platform within the same instrument run. Forensic DNA markers are "weighed" with sufficient accuracy to provide base compositions (the number of A's, G's, C's and T's). Base compositions for multiple makers provide a DNA profile for an individual that can be referenced to existing forensics databases.

Mitochondrial profiling is not hindered by heteroplasmy, is capable of resolving mixtures, and mtDNA profiles may be compared to existing sequence databases or relevant sequence data may be compared to a MS-derived profile database. More than 100,000 mtDNA profiles have been derived using the Ibis platform allowing an unprecedented ability to interrogate the prevalence and extent of genetic variation in the non-coding region, including length and point heteroplasmy.

The PLEX-ID system is capable of identifying and storing polymorphic STR and Y-STR alleles while maintaining backwards-compatibility with current STR databases. Because an intrinsic property of each amplification product (i.e., its mass) is measured, allelic ladders are unnecessary for STR analysis. For autosomal STR loci, polymorphisms have been observed in 11 of 13 core CODIS loci, with six loci displaying polymorphisms in >20% of observed alleles observed (D13S317, D21S11, D3S1358, D5S818, D8S1179 and vWA). Moreover, instances of heterozygous loci displaying alleles of the same length (one or both alleles being polymorphic) have been observed in 9 of the 13 core CODIS loci, with five loci (D13S317, D3S1358, D5S818, D8S1179 and vWA) displaying frequencies of >5% of individuals being heterozygous with same-length alleles.

Several interesting applications of the Abbott MS platform will be discussed.