

## SNP AND STR PROFILING OF FORENSIC SAMPLE TYPES

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SNP genotyping in conjunction with autosomal STR profiling is an important tool for the analysis of difficult samples, particularly in evidence cases where the DNA is highly degraded or in paternity cases where mutations at STR loci complicate genotype interpretation. SNPs are the most abundant genetic variations in the human genome and thus provide a wealth of informative markers for use in human identification (HID) applications. SNPs are genetically stable, with an equal background of forward and back mutation rate and can be genotyped using very short DNA fragments (40 – 60 bp). Major constraints in the development of SNP genotyping systems have been their sensitivity and the ease-of-use of detection platforms. We have developed the GenPlex™ HID system, which offers speed, flexibility, and high quality results for forensic applications. This system works by combining multiplexed PCR amplification of SNP containing genomic target regions with a method that detects polymorphisms within amplicons using an oligonucleotide ligation assay (OLA). The GenPlex™ HID system has been optimized to detect up to 48 SNPs in a single reaction. The 48-plex SNP genotyping system provides both high quality results and a high power of discrimination. Furthermore, more than 90 DNA samples can be analyzed simultaneously.

We demonstrate the utility of SNP typing using a variety of sample types including blood stains on denim and cotton cloth, samples spiked with PCR inhibitors, saliva on swabs, semen on cotton fabric, bones, teeth, chewing gum, cigarette butts, and tape lifts. The DNA was extracted using the PrepFiler™ Forensic DNA Extraction kit and samples were processed for STR profiling using the Identifiler® and MiniFiler™ kits and for SNP typing using the GenPlex™ HID system. SNP analysis increased the probative value of the evidence in degraded samples where STR profiles were either partial or inconclusive demonstrating the importance of using two independent genotyping systems, e.g. autosomal STRs and SNPs, in forensic DNA analysis.