Single Nucleotide Polymorphisms and Microarray Technology in Forensic Genetics — Development and Application to Mitochondrial DNA

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ABSTRACT: Variations in the genome, due to base substitutions, insertions, or deletions at single positions, are known as single nucleotide polymorphisms (SNPs). Approximately 85% of human variation is based on such polymorphisms. Therefore, there is an abundance of human SNPs that are available for forensic identity testing purposes. SNP analyses also may be suitable for some forensic identity cases, because they can be detected in small-sized amplicons, allowing for genetic analysis of substantially degraded DNA. While SNP analysis is unlikely to replace short tandem repeat loci typing for routine casework, SNPs may prove useful for certain circumstances, for example, typing mitochondrial DNA (mtDNA). Although sequencing mtDNA enables detection of all SNPs contained within the region of interest, it is currently not a practical approach for simultaneously typing SNPs that reside throughout the entire mtDNA genome. A variety of alternate methods to detect SNPs are available that may facilitate mtDNA analysis. All the methods include amplification, typically by the polymerase chain reaction, of the region containing the SNP of interest. Most assays are based on either hybridization of a probe to amplified product or primer extension chemistry, and multiplexing is possible. Some of these methodologies are: chips, SNaP shot™, Luminex 100™, SNPstream® UHT, and Pyrosequencing™. SNP analysis of mtDNA, both in the noncoding and coding regions, has been demonstrated using a number of these formats.

KEY WORDS: Chips, forensic science, identity testing, Luminex 100™, mitochondrial DNA, SNaP shot™, SNP, SNPstream® UHT, PCR, Pyrosequencing™.