Preface

At present, most of the validated forensic DNA profiling procedures are based on high-resolution and high-throughput capillary electrophoresis separation and detection systems of PCR amplicons obtained from DNA genomic markers with different inheritance patterns (autosomal, Y-chromosome or X-chromosome linked, and mitochondrial DNA). A high degree of standardization has been achieved in the field of typing procedures for PCR products obtained with fluorogenic labeled primers, separated by capillary electrophoresis, and detected by the fluorescence emission induced by laser on a CCD camera. Current systems have acquired a high degree of accuracy and precision in the identification of allelic variants (with a resolution of one base), and also an improvement in the sensitivity of PCR amplicon detection (with reproducible signals even from picograms of DNA template). Another advantage of current capillary electrophoresis systems (with 5–6 multichannel detection technology) is a greater capacity for simultaneous detection of multiple markers. It can be detected up to 15–26 short tandem repeats (STRs) multiallelic markers, perform simultaneously about 50 biallelic single nucleotide polymorphisms (SNPs) or detect 4-dye-terminator sequencing reactions in a single capillary electrophoresis injection.

Human forensic identification is currently performed using commercial kits for STR multiplexing. Basically there are two sets of standardized STR markers in the criminal DNA databases around the world: the European standard set of 12 STR markers and the USA CODIS standard of 13 STR markers. They form together a standard of 18 STR markers in total. Development of mini-STR markers and their recent incorporation into commercial kits have improved the application of these markers in severely degraded DNA samples.

Other source of human genetic variation used in forensic casework is the analysis of sex chromosome markers. The Y-chromosome STR haplotypes are of special interest in the specific analysis of the male component of DNA mixtures that are very frequently faced by forensic labs in sexual assault cases. Another application of Y-STR haplotyping using a commercial multiplex PCR system of 17 loci, is the identification of human remains and missing persons checked against the appropriate reference samples of paternal relatives. As regards X-chromosome STR analysis, it provides complementary analysis in some kinship deficiency cases.

The automated sequence analysis of mitochondrial DNA hypervariable regions is especially important in the analysis of forensic samples negative for nuclear DNA, or when it is necessary to identify human remains by comparing them with maternal relatives.

Multiplex PCR-CE assays of autosomal SNP markers have been developed for human individualization of degraded samples and as a complementary tool in paternity testing. The analysis of autosomal, Y-chromosome, and mtDNA SNPs with well-differentiated allele or haplotype frequencies among global population-groups has been also applied to the inference of likely ancestry.

DNA Electrophoresis Protocols for Forensic Genetics offers a comprehensive coverage of the most modern current electrophoretic protocols and interpretation guidelines used in forensic genetics for the analysis of amplified human DNA fragments and DNA sequencing.

It includes protocols for profiling of autosomal STRs (see Chapters 1–3, and 13–15), Y-STRs (see Chapter 4), X-STRs (see Chapter 5), autosomal SNPs and INDELS (see Chapters 6–8, 10), Y-SNPs (see Chapter 9), mtDNA-SNPs (see Chapter 11), and mtDNA hypervariable regions HV1 and HV2 (see Chapters 19–21).

Besides the use of DNA electrophoresis on different applications for human identification (criminal investigations, kinship analysis, degraded samples, low template DNA, etc.) the book also covers some interesting electrophoretic protocols for molecular identification of nonhuman species with interest in forensic botany (see Chapters 17 and 18), forensic veterinary (see Chapters 16, 22, and 23), and microbial forensics (see Chapter 26).

Finally the book explores novel forensic applications of capillary electrophoresis to target messenger RNA markers for body fluid identification (see Chapter 12), and some forensic applications of microchip capillary electrophoresis (see Chapters 24 and 25).

DNA Electrophoresis Protocols for Forensic Genetics has been written by highly recognized professionals and specialists from different forensic DNA laboratories around the world dealing with casework and using validated technology and high quality standards. This book was made possible, thanks to them.

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