Preface

The discovery of polymorphisms in repetitive DNA by Dr. Alec Jeffreys and coworkers in 1985 has had a tremendous impact on forensic genetics. Since then we have witnessed a revolution in the field of forensic identification, and different markers and technologies for DNA typing have moved at a breathtaking pace.

Rapid advances in technology, from serological or electrophoretic analysis of protein polymorphisms to direct investigation of the underlying DNA polymorphisms, occurred in a very short space of time in the mid-1980s. Consequently, the incorporation of modern molecular biological techniques in the forensic genetic laboratory has resulted in major benefits for justice.

DNA analysis has become the standard method applied by most forensic genetic labs, especially in criminal forensic casework (e.g., analysis of stains and hairs, identification of human remains, and paternity testing). Polymerase chain reaction (PCR)-based DNA typing systems have made it possible to analyze DNA obtained from only a few cells as well as from highly degraded human samples (recently demonstrated by the identification of relatively old human remains). The potential of DNA typing has made possible the resolution of immigration problems and complicated paternity testing cases when the father is not available. Rapid identification of individuals in mass disaster using DNA typing has also been possible. Computerized DNA databases for the identification of criminal offenders have been created in some countries.

Owing to these many impressive applications, the media have taken great interest in DNA profiling, mainly because of the value of the evidence presented through DNA profiling in certain well-known legal cases.

Initially, the use of DNA profiling was very controversial in some countries, perhaps because of a hasty introduction of this new methodology. Ironically, however, this has contributed to a much more reliable use of DNA profiling.

Two parallel upheavals concerning the introduction of DNA typing technology have been accountable for the aforementioned: the introduction of quality control and accreditation schemes and, in particular, the spreading use of the statistics in the evaluation of DNA evidence. Also, progress in standardizing the tests has proven even more important than the technical advances.

In addition to the DNA revolution, the evolution and development of DNA markers and technologies themselves have been rapid and spectacular. In only a few years we have progressed from the original multilocus DNA fingerprint

analysis of DNA minisatellites, through single locus probe analysis of specific minisatellites, to a host of systems based on the PCR technique.

Microsatellites or short-tandem repeats (STRs) have been almost completely substituted for minisatellites in forensic labs. STRs were first analyzed in manual electrophoretic systems. The introduction of fluorescent-based technology and the use of DNA sequencers have revolutionized the field, allowing the typing of large multiplexes, as well as the automation of the typing procedure. Commercially available and robust multiplexes with up to 15 STRs are routinely used by most of the forensic labs.

But new markers and methods of detection have been proposed, and the most important new advances are the introduction of the use of mtDNA and Y chromosome polymorphisms and especially the new use of single nucleotide polymorphisms (SNPs). It is now clear that SNP typing will be of prime importance in the field, owing to the potential advantages of this type of marker, especially for the analysis of degraded samples.

Because STR typing is familiar in all the forensic labs and the typing protocols are well established, we have decided to focus *Forensic DNA Typing Protocols* on the newer methods and technologies forensic scientists use to solve certain types of cases and to implement these new DNA typing methods in their laboratories. In addition, we have included a chapter on how to create large STR multiplexes, since some labs are interested in the design of STR multiplexes for specific purposes (e.g., STRs with short amplicons for degraded samples; pentanucleotide STRs for the analysis of mixtures).

Forensic DNA Typing Protocols provides protocols for the major methods of DNA analysis that have been recently introduced for identity testing, including Y chromosome, mtDNA, and SNP typing. Chapters with protocols for new applications in the forensic genetics labs—such as species identification or typing of CYP polymorphisms for the analysis of adverse reactions to drugs—have also been included. Ancient DNA is another field of forensic and anthropological interest where there is a need for well-tested protocols from laboratories with extensive experience in the field; two chapters are devoted to this topic. Finally, proper DNA quantification is a crucial requirement for the analysis of critical forensic samples, including mixtures, and new methods based on real-time PCR are now available. For this reason two chapters including protocols for DNA quantification of forensic samples and for the determination of the number of amelogenin gene copies have been added.

I am indebted to Professor John Walker, the series editor, for his helpful encouragement and patience, but I especially owe my thanks to all the contributing authors who have made this book possible.

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