

Contents

Preface	ix
About the Authors	x
1 Introduction to forensic genetics	1
Forensic genetics	1
A brief history of forensic genetics	2
References	5
2 DNA structure and the genome	7
DNA structure	7
Organization of DNA into chromosomes	7
The structure of the human genome	9
Genetic diversity of modern humans	11
The genome and forensic genetics	11
Tandem repeats	12
Single nucleotide polymorphisms (SNPs)	13
Further reading	14
References	14
3 Biological material – collection, characterization and storage	17
Sources of biological evidence	17
Collection and handling of material at the crime scene	19
Identification and characterization of biological evidence	19
Evidence collection	20
Sexual and physical assault	21
Presumptive testing	21
Storage of biological material	23
References	24
4 DNA extraction and quantification	27
DNA extraction	27
General principles of DNA extraction	27
DNA extraction from challenging samples	30
Quantification of DNA	32
DNA IQ™ system	36
References	36

5	The polymerase chain reaction	39
	The evolution of PCR-based profiling in forensic genetics	39
	DNA replication – the basis of the PCR	40
	The components of PCR	40
	The PCR process	42
	PCR inhibition	44
	Sensitivity and contamination	45
	The PCR laboratory	46
	Further reading	48
	References	48
6	The analysis of short tandem repeats	51
	Structure of STR loci	51
	The development of STR multiplexes	51
	Detection of STR polymorphisms	54
	Interpretation of STR profiles	56
	Further reading	61
	References	61
7	Assessment of STR profiles	65
	Stutter peaks	65
	Split peaks (+/–A)	65
	Pull-up	67
	Template DNA	68
	Overloaded profiles	68
	Low copy number DNA	68
	Peak balance	70
	Mixtures	70
	Degraded DNA	71
	References	73
8	Statistical interpretation of STR profiles	75
	Population genetics	75
	Deviation from the Hardy–Weinberg equilibrium	76
	Statistical tests to determine deviation from the Hardy–Weinberg equilibrium	77
	Estimating the frequencies of STR profiles	78
	Corrections to allele frequency databases	78
	Which population frequency database should be used?	83
	Conclusions	83
	Further reading	84
	References	84

9	The evaluation and presentation of DNA evidence	87
	Hierarchies of propositions	87
	Likelihood ratios	89
	Two fallacies	93
	Comparison of three approaches	94
	Further reading	95
	References	95
10	Databases of DNA profiles	97
	The UK National DNA database (NDNAD)	97
	International situation	102
	References	104
11	Kinship testing	105
	Paternity testing	105
	Identification of human remains	111
	Further reading	112
	References	112
12	Single nucleotide polymorphisms	115
	SNPs – occurrence and structure	115
	Detection of SNPs	115
	SNP detection for forensic applications	117
	Forensic applications of SNPs	119
	SNPs compared to STR loci	120
	Further reading	121
	References	121
13	Lineage markers	125
	Mitochondria	125
	Applications of mtDNA profiling	127
	The Y chromosome	130
	Forensic applications of Y chromosome polymorphisms	131
	Further reading	132
	References	133
	Appendix 1 Forensic parameters	137
	Appendix 2 Useful web links	139
	Glossary	141
	Abbreviations	145
	Index	147