

Contents

Sections marked † are at a higher level of specialization and/or are peripheral topics not required to follow the remainder of the book. They may be skipped at first reading.

Preface to the second edition	xi
Preface to the first edition	xiii
1 Introduction	1
1.1 Weight-of-evidence theory	1
1.2 About the book	3
1.3 DNA profiling technology	4
1.4 What you need to know already	4
1.5 Other resources	5
2 Crime on an island	7
2.1 Warm-up examples	7
2.1.1 <i>People v. Collins</i> (California, 1968)	7
2.1.2 Disease testing: positive predictive value (PPV)	8
2.1.3 Coloured taxis	10
2.2 Rare trait identification evidence	10
2.2.1 The ‘island’ problem	11
2.2.2 A first lesson from the island problem	12
2.3 Making the island problem more realistic	13
2.3.1 The effect of uncertainty about p	13
2.3.2 Uncertainty about N	14
2.3.3 The effect of possible typing errors	15
2.3.4 The effect of searches	16
2.3.5 The effect of other evidence	17
2.3.6 The effects of relatives and population subdivision	18
2.4 Weight-of-evidence exercises	19
3 Assessing evidence using likelihoods	21
3.1 Likelihoods and their ratios	21
3.2 The weight-of-evidence formula	23
3.2.1 The population \mathcal{P}	23

3.2.2	Grouping the R_X	24
3.2.3	Application to the island problem	25
3.3	General application of the formula	25
3.3.1	Several items of evidence	25
3.3.2	The role of the expert witness	28
3.4	Consequences for DNA evidence	28
3.4.1	Many possible culprits	28
3.4.2	Incorporating the non-DNA evidence	30
3.4.3	Relatives	31
3.4.4	Laboratory and handling errors	31
3.4.5	Database searches	33
3.5	Derivation of the weight-of-evidence formula †	35
3.5.1	Bayes' theorem	35
3.5.2	Uncertainty about p and N	35
3.5.3	Grouping the alternative possible culprits	37
3.5.4	Typing errors	37
3.6	Further weight-of-evidence exercises	38
4	Profiling technologies	41
4.1	STR typing	41
4.1.1	Anomalies	44
4.1.2	Contamination	47
4.1.3	Low-template DNA (LTDNA) profiling	47
4.2	mtDNA typing	48
4.3	Y-chromosome markers	49
4.4	X-chromosome markers †	50
4.5	SNP profiles †	50
4.6	Sequencing †	52
4.7	Methylation †	52
4.8	RNA †	53
4.9	Fingerprints †	53
5	Some population genetics for DNA evidence	55
5.1	A brief overview	55
5.1.1	Drift	55
5.1.2	Mutation	58
5.1.3	Migration	59
5.1.4	Selection	59
5.2	F_{ST} or θ	61
5.2.1	Population genotype probabilities	62
5.3	A statistical model and sampling formula	63
5.3.1	Diallelic loci	63
5.3.2	Multi-allelic loci	66
5.4	Hardy–Weinberg equilibrium	68
5.4.1	Testing for deviations from HWE †	69
5.4.2	Interpretation of test results	73

5.5	Linkage equilibrium	73
5.6	Coancestry †	75
5.6.1	One allele	75
5.6.2	Two alleles	75
5.6.3	Three alleles	76
5.6.4	General proof via recursion	76
5.7	Likelihood-based estimation of F_{ST} †	77
5.8	Population genetics exercises	78
6	Inferences of identity	80
6.1	Choosing the hypotheses	80
6.1.1	Post-data equivalence of hypotheses	82
6.2	Calculating LR _s	83
6.2.1	The match probability	83
6.2.2	Single locus	85
6.2.3	Multiple loci: the ‘product rule’	86
6.2.4	Relatives of Q	88
6.2.5	Confidence limits †	90
6.2.6	Other profiled individuals	91
6.3	Application to STR profiles	92
6.3.1	Values for the p_j	92
6.3.2	The value of F_{ST}	94
6.3.3	Choice of population	95
6.3.4	Errors	95
6.4	Application to haploid profiles	96
6.4.1	mtDNA profiles	96
6.4.2	Y-chromosome markers	98
6.5	Mixtures	98
6.5.1	Visual interpretation of mixed profiles	98
6.5.2	Likelihood ratios under qualitative interpretation	99
6.5.3	Quantitative interpretation of mixtures	105
6.6	Identification exercises	106
7	Inferring relatedness	108
7.1	Paternity	108
7.1.1	Weight of evidence for paternity	108
7.1.2	Prior probabilities	109
7.1.3	Calculating LR _s	110
7.1.4	Multiple loci: the effect of linkage	114
7.1.5	Q may be related to c but not the father	115
7.1.6	Incest	116
7.1.7	Mother unavailable	117
7.1.8	Mutation	117
7.2	Other relatedness between two individuals	121
7.2.1	Only the two individuals profiled	121

7.2.2	Profiles of known relatives also available †	122
7.2.3	Software for relatedness analyses	123
7.3	Familial search	125
7.4	Inference of ethnicity †	126
7.5	Inference of phenotype †	128
7.6	Relatedness exercises	128
8	Low-template DNA profiles	130
8.1	Background	130
8.2	Stochastic effects in LTDNA profiles	132
8.2.1	Drop-out	132
8.2.2	Drop-in	133
8.2.3	Peak imbalance	133
8.2.4	Stutter	134
8.3	Computing likelihoods	134
8.3.1	Single contributor allowing for drop-out	134
8.3.2	Profiled contributors not subject to drop-out	135
8.3.3	Modelling drop-in	136
8.3.4	Multi-dose drop-out and degradation	136
8.3.5	Additional contributors subject to drop-out	138
8.3.6	Replicates	138
8.3.7	Using peak heights	139
8.4	Quality of results	141
9	Introduction to likeLTD †	143
9.1	Installation and example R script	143
9.1.1	Input	144
9.1.2	Allele report	145
9.1.3	Arguments and optimisation	145
9.1.4	Output report	147
9.1.5	Genotype probabilities	148
9.2	Specifics of the package	151
9.2.1	The parameters	151
9.2.2	Key features of likeLTD	151
9.2.3	Maximising the penalised likelihood	152
9.2.4	Computing time and memory requirements	153
9.3	Verification	154
10	Other approaches to weight of evidence	157
10.1	Uniqueness	157
10.1.1	Analysis	158
10.1.2	Discussion	159
10.2	Inclusion/exclusion probabilities	160
10.2.1	Identification: single contributor	160
10.2.2	Identification: multiple contributors	161

10.2.3 Paternity	161
10.2.4 Discussion	162
10.3 Hypothesis testing †	162
10.4 Other exercises	163
11 Some issues for the courtroom	165
11.1 The role of the expert witness	165
11.2 Bayesian reasoning in court	166
11.3 Some fallacies	167
11.3.1 The prosecutor's fallacy	167
11.3.2 The defendant's fallacy	168
11.3.3 The uniqueness fallacy	169
11.4 Some UK appeal cases	169
11.4.1 Deen (1993)	169
11.4.2 Adams (1996)	170
11.4.3 Doheny/Adams (1996)	171
11.4.4 Watters (2000)	173
11.4.5 T (2010)	174
11.4.6 Dlugosz (2013)	175
11.5 US National Research Council reports	177
11.6 Prosecutor's fallacy exercises	178
Solutions to exercises	179
References	196
Index	211