

CONTENTS

Preface.....	ix
Biography.....	xiii
Chapter 1 Prerequisites	1
1.1 Genetics.....	1
1.2 Pedigrees	2
1.3 R and the ped suite	2
1.4 Datasets	5
Chapter 2 Pedigrees and Marker Data	7
2.1 Creating Pedigrees	8
2.2 Markers	14
2.3 Loading Pedigree Data From Files.....	20
Exercises	23
Chapter 3 Coefficients of Relatedness	25
3.1 Identity by Descent	26
3.2 Inbreeding Coefficients	27
3.3 Kinship Coefficients	29
3.4 IBD Coefficients of Noninbred Individuals	30
3.5 Identity Coefficients	37
3.6 Constructibility of Kappa.....	39
Exercises	41
Chapter 4 Realised Relatedness	43
4.1 Recombination	45
4.2 The ibdsim2 Package	49
4.3 Variation in Realised Coefficients	53

4.4 Distributions of IBD Segments	56
4.5 The Probability of No IBD Sharing.....	59
Exercises	61
Chapter 5 Probabilities on Pedigrees	63
5.1 Computing Pedigree Likelihoods.....	64
5.2 Factors Affecting Performance.....	68
5.3 Likelihoods With Linked Markers	70
5.4 Modelling Mutations	73
5.5 Modelling Deviation From HWE	77
Exercises	80
Chapter 6 Kinship Testing	83
6.1 Theory and Methods.....	84
6.2 Paternity Testing.....	87
6.3 A Relationship Riddle.....	90
6.4 Missing Person Identification	94
6.5 Power Analysis for Kinship Testing	98
6.6 Dealing With Mutations	101
Exercises	103
Chapter 7 Inference of Pairwise Relatedness	105
7.1 Maximum-Likelihood Estimation of Kappa	106
7.2 Estimation of Identity Coefficients.....	109
7.3 ML Estimation in forrel	109
7.4 Quality Control of Pedigree Data.....	112
7.5 Violating the Assumptions	114
Exercises	117
Chapter 8 Pedigree Reconstruction	119
8.1 Reconstruction by Pairwise Inference.....	120
8.2 Maximum-Likelihood Pedigree Reconstruction	124
Exercises	131

Chapter 9 Linkage Analysis ..

9.1 Theoretical Background ..

9.2 LOD Scores in paramlink ..

9.3 A Case Study

Exercises

Chapter 10 Segregation Analy ..

10.1 Background

10.2 The Bayes Factor

10.3 Computation With the seg ..

10.4 A Case Study

Exercises

Bibliography

Chapter 9 Linkage Analysis	135
9.1 Theoretical Background	136
9.2 LOD Scores in paramlink2	143
9.3 A Case Study	146
Exercises	153
 Chapter 10 Segregation Analysis for Variant Interpretation	 155
10.1 Background	156
10.2 The Bayes Factor	157
10.3 Computation With the segregatr Package	161
10.4 A Case Study	162
Exercises	164
 Bibliography	 167