

Contents

Preface	1
An introduction to human genetics	6
1.1 Some Very Useful Numbers for human population genetics.	7
1.2 A genome owner’s starter pack.	8
1.3 Human genome variation and why it matters.	30
1.4 DNA sequencing: a fundamental tool for studying biology.	47
1.5 Mutation: The ultimate source of genetic variation.	57
Population genetics: the forces that shape genetic variation	73
2.1 Genetic Drift: What happens to alleles over time?	74
2.2 More on genetic drift: The coalescent.	85
2.3 Linkage, recombination, and LD.	104
2.4 Genetic drift in structured populations.	123
2.5 Natural selection: I. Background and models	136
2.6 Natural selection: II. Positive selection and adaptation	150
2.7 Natural Selection III. Genome-wide extent of selection	164
Human population history and structure [outline]	177
3.1 Population structure and ancestry estimation.	178
3.2 Inferring human prehistory from genetic data.	178
3.3 Digging deeper into human history: Ancient DNA.	178
Genetics of phenotypic variation and disease [outline]	179
4.1 A starter pack for human trait genetics	180
4.2 Major effect mutations: monogenic traits	180
4.3 Major effect mutations: somatic mutations and cancer	180
4.4 Complex traits: I. Quantitative genetics	180
4.5 Complex traits: II. The GWAS paradigm	180
4.6 Complex traits: III. More about GWAS	180
4.7 Complex traits: IV. Functional genomics of complex traits	180
4.8 Complex traits: V. stabilizing selection, drift, and adaptation	180
Notes and References	181