

## Part 4.

# Genetics of phenotypic variation and disease

*In the previous sections of the book we have discussed the types of genetic variation, the population genetics forces that act on variation, and how human history and structure shape geographic patterns of variation.*

*In Part 4 we now turn our attention to how **genetic variation shapes phenotypic variation**. We'll cover three main categories of traits: monogenic diseases, cancer, and complex traits, with particular emphasis on complex traits.*

*As you read you should pay attention to the themes that repeat, but with key differences, across the different categories:*

- *The number, allele frequencies and molecular mechanisms of variants;*
- *The types of selection that are most relevant;*
- *The study designs used to identify causal genes and variants;*
- *The main conceptual approaches to data analysis, and major insights.*

*Specifically, we cover the following:*

*Chapter 4.1: A **Starter Pack** of trait genetics: an introduction to the topics in this section.*

*Chapter 4.2: The **genetics of monogenic diseases**: mapping approaches, the major mechanisms, and selection.*

*Chapter 4.3: The **genetics of cancer**, emphasizing aspects of this huge field that intersect our main themes including somatic mutation and selection.*

*Chapter 4.4: **Quantitative Genetics**: statistical models for the inheritance of polygenic traits, including heritability and artificial selection.*

*Chapter 4.5–4.8: An overview of the main approaches for studying **complex traits**, and major emerging themes: GWAS; SNP heritability; regulatory genomics and the mechanisms of variant effects; and polygenic scores.*

*Chapter 4.9: We close with more on the **population genetics of complex traits** including stabilizing selection and polygenic adaptation.*