Part 1.

An introduction to human genetics

In this first part of the book we cover a collection of topics that will be useful background throughout. This includes

- Chapter 1.1: A short list of helpful numbers to guide quantitative thinking about many questions in human genetics.
- Chapter 1.2: An overview of essential principles in genetics, and how these relate to the human genome. Some of this will already be familiar to many readers, but the genomic perspective is not usually covered in this way in introductory classes.
- Chapter 1.3: An **introduction to human genetic variation**: the types of variation, how we quantify variation, and an introduction to how variation affects phenotype. Understanding genetic variation will be the central theme of this book.
- Chapter 1.4: **DNA sequencing** is the fundamental tool for studying genomes, and it's important to understand basic principles about the types of sequencing and the types of data we can collect.
- Chapter 1.5: All genetic variation arose in the past through **mutation**. We provide an overview of mutational processes and rates, emphasizing topics that are relevant for human genetics.