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Chapter **1**

Welcome to Genetics: Just About Everyone Loves a Good Pair of Genes

Welcome to the complex and fascinating world of genetics. Genetics is all about physical traits and the DNA code that supplies the building plans for any organism. This chapter defines the field of genetics and explains what geneticists do. You get an introduction to the big picture and a glimpse at some of the details found in other chapters of this book.

Unwrapping Life's Secrets

Take a moment to think about the vast diversity of living things in the world around you. The instructions to build all of those diverse forms — from a long, limbless snake to a tentacled octopus to a tall redwood tree — are encoded in the chemical pattern of DNA. *Genetics* is the field of science that examines how DNA determines traits and how these traits are passed from one generation to the next. Simply put, genetics affects *everything* about *every* living thing on earth. An organism's *genes* are segments of DNA (deoxyribonucleic acid) that are the fundamental units of heredity. Genes play an essential role in how the organism looks, behaves, and reproduces. Because all biology depends on genes, genetics is a critical foundation for all other life sciences, including agriculture and medicine.



HISTORICAL
STUFF

From a historical point of view, genetics is still a young science. The principles that govern inheritance of traits by one generation from another were described (and promptly lost) less than 150 years ago. Around the turn of the 20th century, the laws of inheritance were rediscovered, an event that transformed biology forever. Even so, it wasn't until the 1950s that the importance of DNA was really understood. Now technology is helping geneticists push the envelope of knowledge every day.

Genetics is generally divided into four major subdivisions. We discuss each of these more in the sections that follow:

- » **Classical, or Mendelian, genetics:** A discipline that describes how physical characteristics (traits) are passed along from one generation to another.
- » **Molecular genetics:** The study of the chemical and physical structures of DNA, its close cousin RNA (ribonucleic acid), and proteins. Molecular genetics also covers how genes do their jobs.
- » **Population genetics:** A division of genetics that looks at the genetic makeup of larger groups.
- » **Quantitative genetics:** A highly mathematical field that examines the statistical relationships between genes and the traits with which they are associated.

In the academic world, many genetics courses begin with classical genetics and proceed through molecular genetics, with a nod to population and quantitative genetics. In general, this book follows the same path, because each division of knowledge builds on the one before it. That said, it's perfectly okay, and very easy, to jump around among disciplines. No matter how you take on reading this book, it provides lots of cross-references to help you stay on track.

Classical Genetics: Studying How Traits Are Transmitted within Families

At its heart, *classical genetics* is the genetics of individuals and their families. It focuses mostly on studying physical traits, or *phenotypes*, as a way to understand the genes behind those traits.



HISTORICAL
STUFF

Gregor Mendel, a monk and part-time scientist, founded the discipline of genetics. Mendel was a gardener with an insatiable curiosity to go along with his green thumb. His observations may have been simple, but his conclusions were jaw-droppingly elegant. This man had no access to technology, computers, or a pocket calculator, yet he determined, with keen accuracy, fundamental laws of inheritance that have stood the test of time.

Classical genetics is sometimes referred to as:

- » **Mendelian genetics:** You start a new scientific discipline, and it gets named after you. Seems fair.
- » **Transmission genetics:** This term refers to the fact that classical genetics describes how traits are passed on, or *transmitted*, from parents to their offspring.

No matter what you call it, classical genetics includes the study of cells and chromosomes, which we cover in Chapters 2 and 6. Cell division is the machine that drives inheritance, but you don't have to understand combustion engines to drive a car, right? Likewise, you can dive straight into simple inheritance in Chapter 3 and work up to more complicated forms of inheritance in Chapter 4 without knowing anything whatsoever about cell division. (Mendel didn't know anything about chromosomes and cells when he figured this whole thing out, by the way.)

The genetics of sex and reproduction are also part of classical genetics. Various combinations of genes and *chromosomes* (strands of DNA) determine biological sex. But the subject of sex gets even more complicated and interesting: The environment plays a role in determining the sex of some organisms (like crocodiles and turtles), and other organisms can even change sex with a change of address. If this has piqued your interest, you can find out all the astonishing details in Chapter 6. (Of note, we use the term *sex* throughout this book instead of the term *gender*. *Sex* is what defines males and females from a biological perspective. A person's *gender*, on the other hand, may also be influenced by social and cultural factors, and may differ from one's biological sex.)

Classical genetics provides the framework for many subdisciplines. The study of chromosome disorders such as Down syndrome, which we cover in Chapter 12, relies on cell biology and an understanding of what happens during cell division. Genetic counseling, which we cover in Chapter 14, also relies on understanding patterns of inheritance to interpret people's medical histories from a genetics perspective. In addition, forensics, covered in Chapter 17, uses Mendelian genetics to determine paternity and to work out who's who with DNA fingerprinting.

Molecular Genetics: Diving into DNA Science

Classical genetics concentrates on studying outward appearances, while the study of actual genes falls under the heady title of *molecular genetics*. The area of operations for molecular genetics includes all the machinery that runs cells and manufactures the structures called for by the plans found in genes. The focus of molecular genetics includes the physical and chemical structures of the double helix, DNA, which is broken down in all its glory in Chapter 5. The messages hidden in your DNA (your genes) constitute the building instructions for your appearance and everything else about you — from how your muscles function and how your eyes blink to your blood type, your susceptibility to particular diseases, and everything in between. How that DNA (and the immense amount of information it contains) is packaged in your cells is covered in Chapter 6, which reviews the structure and function of chromosomes.

Your genes are expressed through a complex system of interactions that begins with *transcription* — copying DNA's messages into a somewhat temporary form called RNA, which is short for *ribonucleic acid* and is covered in Chapter 8. RNA carries the DNA message through the process of *translation*, covered in Chapter 9, which in essence is like taking a blueprint to a factory to guide the manufacturing process. Where your genes are concerned, the factory makes the proteins (from the RNA blueprint) that get folded in complex ways to make the various components of the cells and tissues in the human body. The study of *gene expression* (how genes get turned on and off, which we review in Chapter 10) and how the genetic code works at the levels of DNA and RNA are considered parts of molecular genetics.

Research on the causes of cancer and the hunt for better treatments, which we address in Chapter 13, focuses on the molecular side of things because tumors result from changes in the DNA, called *mutations*. Chapter 11 covers mutations in detail. Gene therapy, covered in Chapter 15, and genetic engineering, covered in Chapter 19, are subdisciplines of molecular genetics.

Population Genetics: Studying the Genetics of Groups

The science of genetics has a strong foundation in math. (In fact, Gregor Mendel studied to be a math teacher.) One area in which calculations are used to describe what goes on genetically is population genetics.



REMEMBER

If you use Mendelian genetics and examine the inheritance patterns of many different individuals who have something in common, like geographic location, you can study population genetics. *Population genetics* is the study of the genetic diversity of a subset of a particular species (for details, you can flip ahead to Chapter 16). Basically, it's a search for patterns that help describe the genetic signature of a particular group, such as the consequences of migration, isolation from other populations, and mating choices.

Population genetics helps scientists understand how the collective genetic diversity of a population influences the health of individuals within the population. For example, cheetahs are lanky cats; they're the speed demons of Africa. Population genetics has revealed that all cheetahs are extremely genetically similar; in fact, they're so similar that a skin graft from one cheetah would be accepted by any other cheetah. Because the genetic diversity of cheetahs is so low, conservation biologists fear that a disease could sweep through the population and kill off all the individuals of the species. It's possible that no animals would be resistant to the disease, and therefore, none would survive, leading to the extinction of this amazing predator.

Evolutionary genetics is a type of population genetics that involves studying how traits change over time. We review evolutionary genetics in Chapter 16. Describing the genetics of populations from a mathematical standpoint is also critical to forensics, as explained in Chapter 17. To pinpoint the uniqueness of one DNA fingerprint, geneticists need to sample the genetic fingerprints of many individuals and decide how common or rare a particular pattern may be. Likewise, medicine uses population genetics to determine how common particular DNA changes are and to develop new medicines to treat disease (discussed in Chapter 15).

Quantitative Genetics: Exploring the Heredity of Complex Traits

Quantitative genetics examines traits that vary in subtle ways and relates those traits to the underlying genetics of an organism. A combination of whole suites of genes and environmental factors controls characteristics like retrieving ability in

dogs, egg size or number in birds, and running speed in humans. Mathematical in nature, quantitative genetics takes a rather complex statistical approach to estimate how much variation in a particular trait is due to the environment and how much is actually genetic.

One application of quantitative genetics is determining how heritable a particular trait is. This measure allows scientists to make predictions about how offspring will turn out based on characteristics of the parent organisms. Heritability gives some indication of how much a characteristic (like seed production) can change when selective breeding (or, in evolutionary time, natural selection) is applied.

GREAT GENETICS WEBSITES TO EXPLORE

The Internet is an unparalleled source of information about genetics. With just a few mouse clicks, you can find the latest discoveries and attend the best courses ever offered on the subject. Here's a quick sample.

- To see a great video that explains genetics and gives it a human face, check out "Cracking the Code of Life": <https://www.pbs.org/wgbh/nova/genome/program.html>.
- New discoveries are unveiled every day. To stay current, log on to www.sciencedaily.com/news/plants_animals/genetics and www.sciencenews.org/topic/genetics.
- For students, <http://learn.genetics.utah.edu> can't be beat. From the basics of heredity to virtual labs to cloning, it's all there in easy-to-grasp animations and language.
- Want to get all the details about genes and diseases? Medline Plus provides straightforward explanations on numerous topics: <https://ghr.nlm.nih.gov>. You could also start at <https://www.ncbi.nlm.nih.gov/books/NBK22183> for a review of the basics. More advanced (and greatly detailed) information is available at Online Mendelian Inheritance in Man (OMIM): www.ncbi.nlm.nih.gov/omim.
- If you're interested in a career in genetics, the American Society for Human Genetics is ready to help: <https://www.ashg.org/careers-learning/career-flowchart>. (You can also check out some career descriptions in Chapter 22.)