Chapter 1

What Genetics Is and Why You Need to Know Some

In This Chapter

- Defining the subject of genetics and its various subdivisions
- Observing the day-to-day activities in a genetics lab
- Getting the scoop on career opportunities in genetics

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 explains what the field of genetics is and 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.

What Is Genetics?

Genetics is the field of science that examines how traits are passed from one generation to the next. Simply put, genetics affects *everything* about *every* living thing on earth. An organism's *genes,* snippets of DNA that are the fundamental units of heredity, control how the organism looks, behaves, and reproduces. Because all biology depends on genes, understanding genetics as a foundation for all other life sciences, including agriculture and medicine, is critical.

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. But even then, the importance of the star of the genetics show, DNA, wasn't really understood until the 1950s. Now, technology is helping geneticists push the envelope of knowledge every day.

Genetics is generally divided into four major subdivisions:

- **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, 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 they encode.

In the academic world, many genetics courses begin with classical genetics and proceed through molecular genetics, with a nod to population, evolutionary, or quantitative genetics. 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, I provide lots of cross references to help you stay on track.

Classical genetics: Transmitting traits from generation to generation

At its heart, *classical genetics* is the genetics of individuals and their families. It focuses mostly on studying physical traits, or *phenotypes,* as a stand-in for the genes that control appearance.

Gregor Mendel, a humble monk and part-time scientist, founded the entire 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, exactly how inheritance works.

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,* by parents to their offspring.

No matter what you call it, classical genetics includes the study of cells and chromosomes (which I delve into in Chapter 2). 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 (see 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 sex, as in maleness and femaleness. 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 I've piqued your interest, you can find out all the slightly kinky details in Chapter 5.

Classical genetics provides the framework for many subdisciplines. Genetic counseling (which I cover in Chapter 12) depends heavily on understanding patterns of inheritance to interpret people's medical histories from a genetics perspective. The study of chromosome disorders such as Down syndrome (see Chapter 15) relies on cell biology and an understanding of what happens during cell division. Forensics (see Chapter 18) also uses Mendelian genetics to determine paternity and to work out who's who with DNA fingerprinting.

Molecular genetics: DNA and the chemistry of genes

REMEMBER

Classical genetics concentrates on studying outward appearances, but 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 I break down in all its glory in Chapter 6. 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.

Your genes are expressed through a complex system of interactions that begins with copying DNA's messages into a somewhat temporary form called RNA (see Chapter 9). RNA carries the DNA message through the process of *translation* (covered in Chapter 10), 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 you.

The study of *gene expression* (how genes get turned on and off; flip to Chapter 11) 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 a cure (which I address in Chapter 14) focuses on the molecular side of things, because changes (referred to as *mutations*) occur at the chemical level of DNA (see Chapter 13 for coverage of mutations). Gene therapy (see Chapter 16), genetic engineering (see Chapter 19), and cloning (see Chapter 20) are all subdisciplines of molecular genetics.

Population genetics: Genetics of groups

Much to the chagrin of many undergrads, genetics is surprisingly mathematical. One area in which calculations are used to describe what goes on genetically is population genetics.

If you take Mendelian genetics and examine the inheritance patterns of many different individuals who have something in common, like geographic location, then you have population genetics. *Population genetics* is the study of the genetic diversity of a subset of a particular species (for details, jump to Chapter 17). In essence, it's a search for patterns that help describe the genetic signature of a particular group, such as the consequences of travel, isolation (from other populations), mating choices, geography, and behavior.

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 very, very 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.

Describing the genetics of populations from a mathematical standpoint is critical to forensics (see Chapter 18). To pinpoint the uniqueness of one DNA fingerprint, geneticists have to sample the genetic fingerprints of many individuals and decide how common or rare a particular pattern may be. Medicine also uses population genetics to determine how common particular mutations are and to develop new medicines to treat disease. For details on mutations, flip to Chapter 13; see Chapter 21 for information on genetics and the development of new medicines. Also, e*volutionary genetics,* or how traits change over time, is new to this edition; I cover the subject in Chapter 17.

Quantitative genetics: Getting a handle on heredity

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.

Living the Life of a Geneticist

Daily life for a geneticist can include working in the lab, teaching in the classroom, and interacting with patients and their families. In this section, you discover what a typical genetics lab is like and get a rundown of a variety of career paths in the genetics field.

Exploring a genetics lab

A genetics lab is a busy, noisy place. It's full of equipment and supplies and researchers toiling away at their workstations (called *lab benches,* even though the bench is really just a raised, flat surface that's conducive to working while standing up). Depending on the lab, you may see people looking very official in white lab coats or researchers dressed more casually in jeans and T-shirts. Every lab contains some or all of the following:

- Disposable gloves to protect workers from chemical exposure and to protect DNA and other materials from contamination.
- Pipettes (for measuring even the tiniest droplets of liquids with extreme accuracy), glassware (for liquid measurement and storage), and vials and tubes (for chemical reactions).
- Electronic balances for making super-precise measurements of mass.
- Chemicals and ultrapure water.

- A refrigerator (set at 40 degrees Fahrenheit), a freezer (at –4 degrees), and an ultracold freezer (at –112 degrees) for storing samples.

  Repeated freezing and thawing causes DNA to break into tiny pieces, which destroys it. For that reason, freezers used in genetics labs aren't frost-free, because the temperature inside a frost-free freezer cycles up and down to melt any ice that forms.

- Centrifuges for separating substances from each other. Given that different substances have different densities, centrifuges spin at extremely high speeds to force materials to separate so that researchers can handle them individually.
- Incubators for growing bacteria under controlled conditions. Researchers often use bacteria for experimental tests of how genes work.
- Autoclaves for sterilizing glassware and other equipment using extreme heat and pressure to kill bacteria and viruses.
- Complex pieces of equipment such as thermocyclers (used for PCR; see Chapter 18) and DNA sequencers (see Chapter 8).
- Lab notebooks for recording every step of every reaction or experiment in nauseating detail. Geneticists must fully replicate (run over and over) every experiment to make sure the results are valid. The lab notebook is also a legal document that can be used in court cases, so precision and completeness are musts.
- Desktop computers packed with software for analyzing results and for connecting via the Internet to vast databases packed with genetic information (flip to the end of this chapter for the addresses of some useful Web sites).

Researchers in the lab use the various pieces of equipment and supplies from the preceding list to conduct experiments and run chemical reactions. Some of the common activities that occur in the genetics lab include

- Separating DNA from the rest of a cell's contents (see Chapter 6).
- Measuring the purity of a DNA sample and determining how much DNA (by weight) is present.
- Mixing chemicals that are used in reactions and experiments designed to analyze DNA samples.
- Growing special strains of bacteria and viruses to aid in examining short stretches of DNA (see Chapter 16).
- Using DNA sequencing (which I cover in Chapter 8) to learn the order of bases that compose a DNA strand (which I explain in Chapter 6).
- Setting up polymerase chain reactions, or PCR (see Chapter 18), a powerful process that allows scientists to analyze even very tiny amounts of DNA.

- Analyzing the results of DNA sequencing by comparing sequences from many different organisms (you can find this information in a massive, publicly available database — see the end of this chapter).
- Comparing DNA fingerprints from several individuals to identify perpetrators or to assign paternity (see Chapter 18).
- Holding weekly or daily meetings where everyone in the lab comes together to discuss results or plan new experiments.

Sorting through jobs in genetics

Whole teams of people contribute to the study of genetics. The following are just a few job descriptions for you to mull over if you're considering a career in genetics.

Lab tech

Lab technicians handle most of the day-to-day work in the lab. The tech mixes chemicals for everyone else in the lab to use in experiments. Techs usually prepare the right sorts of materials to grow bacteria (which are used as carriers for DNA; see Chapter 16), set up the bacterial cultures, and monitor their growth. Techs are also usually responsible for keeping all the necessary supplies straight and washing the glassware — not a glamorous job but a necessary one, because labs use tons of glass beakers and flasks that have to be cleaned.

When it comes to actual experiments, lab technicians are responsible for separating the DNA from the rest of the tissue around it and testing it for purity (to make sure no contaminants, like proteins, are present). Using a rather complicated machine with a strong laser, the tech can also measure exactly how much DNA is present. When a sufficiently pure sample of DNA is obtained, techs may analyze the DNA in greater detail (with PCR or sequencing reactions).

The educational background needed to be a lab tech varies with the amount of responsibility a particular position demands. Most techs have a minimum of a bachelor's degree in biology or some related field and need some background in microbiology to understand and carry out the techniques of handling bacteria safely and without contaminating cultures. And all techs must be good record-keepers, because every single activity in the lab must be documented in writing in the lab notebook.

Graduate student and post-doc

At most universities, genetics labs are full of *graduate students* working on either master's degrees or PhDs. In some labs, these students may be carrying out their own, independent research. On the other hand, many labs focus their work on a specific problem, like some specialized approach to studying cancer, and every student in that sort of lab works on some aspect of what his or her professor studies. Graduate students do a lot of the same things that lab techs do (see the preceding section), as well as design experiments, carry out those experiments, analyze the results, and then work to figure out what the results mean. Then, the graduate student writes a long document (called a *thesis* or *dissertation*) to describe what was done, what it means, and how it fits in with other people's research on the subject. While working in the lab, grad students take classes and are subjected to grueling exams (trust me on the grueling part).

All graduate students must hold a bachelor's degree. Performance on the standardized GRE (Graduate Record Exam) determines eligibility for admission to master's programs and may be used for selection for fellowships and awards.

TIP

If you're going to be staring down this test in the near future, you may want to get a leg up by checking out *The GRE Test For Dummies,* by Suzee Vlk, Michelle Rose Gilman, and Veronica Saydak (Wiley).

In general, it takes two or three years to earn a master's degree. A doctorate (denoted by *PhD*) usually requires anywhere from four to seven years of education beyond the bachelor's level.

After graduating with a PhD, a geneticist-in-training may need to get more experience before hitting the job market. Positions that provide such experience are collectively referred to as *post-docs* (post-doctoral fellows). A person holding a post-doc position is usually much more independent than a grad student when it comes to research. The post-doc often works to learn new techniques or to acquire a specialty before moving on to a position as a professor or a research scientist.

Research scientist

Research scientists work in private industries, designing experiments and directing the activities of lab techs. All sorts of industries employ research scientists, including

- Pharmaceutical companies, to conduct investigations on how drugs affect gene expression (see Chapter 11) and to develop new treatments such as gene therapy (see Chapter 16).
- Forensics labs, to analyze DNA found at crime scenes and to compare DNA fingerprints (see Chapter 18).
- Companies that analyze information generated by genome projects (human and others; see Chapter 11).
- Companies that support the work of other genetics labs by designing and marketing products used in research, such as kits used to run DNA fingerprints.

A research scientist usually holds a master's degree or a PhD. With only a bachelor's degree, several years of experience as a lab tech may suffice. Research scientists have to be able to design experiments and analyze results using statistics. Good record-keeping and strong communication skills (especially in writing) are musts. Most research scientists also have to be capable of managing and supervising people. In addition, financial responsibilities may include keeping up with expenditures, ordering equipment and supplies, and wrangling salaries of other personnel.

College or university professor

Professors do everything that research scientists do with the added responsibilities of teaching courses, writing proposals to get funds to support research, and writing papers on their research results for publication in reputable, peer-reviewed journals. Professors also supervise the lab techs, graduate students, and post-docs who work in their labs, which entails designing research projects and then ensuring that the projects are done correctly in the right amount of time (and under budget!).

Small schools may require a professor to teach as many as three courses every semester. Upper-tier institutions (think Big Ten or Ivy League) may require only one course of instruction per year. Genetics professors teach the basics as well as advanced and specialty courses like recombinant DNA (see Chapter 16) and population genetics (see Chapter 17).

To qualify for a professorship, universities require a minimum of a PhD, and most require additional post-doctoral experience. Job candidates must have already published research results to demonstrate the ability to do relevant research. Most universities also look for evidence that the professor-to-be will be successful at getting grants, which means the candidate must usually land a grant before getting a job.

Genetic counselor

Genetic counselors work with medical personnel to interpret the medical histories of patients and their families. The counselor usually works directly with the patient to assemble all the information into a family tree (see Chapter 12) and then looks for patterns to determine which traits may be hereditary. Counselors can also tell which diseases a patient is most likely to inherit. Genetic counselors are trained to conduct careful and thorough interviews to make sure that no information is missed or left out.

Genetic counselors usually hold a master's degree. Training includes many hours working with patients to hone interview and analysis skills (under the close supervision of experienced professionals, of course). The position requires excellent record-keeping skills and strict attention to detail. Genetic counselors also have to be good at interacting with all kinds of people, including research scientists and physicians. And the ability to communicate very well, both in writing and verbally, is a must.

The most essential skill of a genetic counselor is the ability to be nonjudgmental and nondirective. The counselor must be able to analyze a family history without bias or prejudice and inform the patient of his or her options without recommending any one course of action over another. Furthermore, the counselor must keep all information about his or her patients confidential, sharing information only with authorized personnel such as the person's own physician in order to protect the patient's privacy.

Great genetics Web sites 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": `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.nature.com/ng/index.html`.
- 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? Start at `www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd` for the basics. You can find more advanced (and greatly detailed) information at Online Mendelian Inheritance in Man: `www.ncbi.nlm.nih.gov/omim/`.
- If you're interested in a career in genetics, the Genetics Society of America is ready to help: `www.genetics-gsa.org/pages/careers_in_genetics.shtml`.