

LEARNING MADE EASY



3rd Edition

Genetics

for
dummies[®]
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Understand cell
biology basics

Explore how genetics
affects your health

Grasp the laws
of inheritance

Tara Rodden Robinson, PhD
Lisa Cushman Spock, PhD, CGC



Genetics

3rd Edition

**by Tara Rodden Robinson, PhD
and Lisa Cushman Spock, PhD, CGC**

**for
dummies[®]**
A Wiley Brand

Genetics For Dummies®, 3rd Edition

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Introduction

Genetics affects all living things. Although sometimes complicated and always diverse, all genetics comes down to basic principles of *heredity* — how traits are passed from one generation to the next — and how DNA is put together. As a science, genetics is a fast-growing field because of its untapped potential — for good and for bad. Despite its complexity, genetics can be surprisingly accessible. Genetics is a bit like peeking behind a movie’s special effects to find a deceptively simple and elegant system running the whole show.

About This Book

Genetics For Dummies, 3rd Edition, is an overview of the entire field of genetics. Our goal is to explain every topic so that anyone, even someone without any genetics background at all, can follow the subject and understand how it works. As in the first and second editions, we include many examples from the frontiers of research. We also make sure that the book has detailed coverage of some of the hottest topics that you hear about in the news, including gene therapy, pharmacogenetics, and gene editing. And we address the practical side of genetics: how it affects your health and the world around you. In short, this book is designed to be a solid introduction to genetics basics and to provide some details on the subject.

Genetics is a fast-paced field; new discoveries are coming out all the time. You can use this book to help you get through your genetics course or for self-guided study. *Genetics For Dummies*, 3rd Edition, provides enough information for you to get a handle on the latest press coverage, understand the genetics jargon that mystery writers like to toss around, and translate information imparted to you by medical professionals. The book is filled with stories of key discoveries and “wow” developments. Although we try to keep things light and inject some humor when possible, we also make every effort to be sensitive to whatever your circumstances may be.

This book is a great guide if you know nothing at all about genetics. If you already have some background, then you’re set to dive into the details of the subject and expand your horizons.

Conventions Used in This Book

It would be very easy for us to use specialized language that you'd need a translator to understand, but what fun would that be? Throughout this book, we try to avoid jargon as much as possible, but at the same time, we use and carefully define terms that scientists actually use. After all, it may be important for you to understand some of these multisyllabic jawbreakers in the course of your studies or your or a loved one's medical treatment.

To help you navigate through this book, we use the following typographical conventions:

- » We use *italic* for emphasis and to highlight new words or terms that we define in the text.
- » We use **boldface** to indicate keywords in bulleted lists or the action parts of numbered steps.
- » We use monospace for websites and email addresses.

Foolish Assumptions

It's a privilege to be your guide into the amazing world of genetics. Given this responsibility, you were in our thoughts often while we were writing this book. Here's how we imagine you, our reader:

- » You're a student in a genetics or biology class.
- » You're curious to understand more about the science you hear reported in the news.
- » You're an expectant or new parent or a family member who's struggling to come to terms with what doctors have told you.
- » You're affected by cancer or some hereditary disease, wondering what it means for you and your family.

If any of these descriptions fit, you've come to the right place.

How This Book Is Organized

We designed this book to cover background material in the first two parts and then all the applications in the rest of the book. We think you'll find it quite accessible.

Part 1: The Lowdown on Genetics: Just the Basics

Part 1 explains how trait inheritance works. The first chapter introduces you to the field of genetics and what genetics professionals may do in their day-to-day work lives. The second chapter gives you a handle on how genetic information gets divided up during cell division; these events provide the foundation for just about everything else that has to do with genetics. From there, we explain simple inheritance of one gene and then move on to more complex forms of inheritance.

Part 2: DNA: The Genetic Material

Part 2 covers what's sometimes called *molecular genetics*. Don't let the word "molecular" scare you off. We give you nitty-gritty details, but we break them down so that you can easily follow along. We track the progress of how your genes work from start to finish: how your DNA is put together, how it gets copied, and how the building plans for your body are encoded in the double helix. To help you understand how scientists explore the secrets stored in your DNA, we also cover how DNA is sequenced. In the process, we relate the fascinating story behind the Human Genome Project.

Part 3: Genetics and Your Health

Part 3 is intended to help you see how genetics affects your health and well-being. We cover the subjects of genetic counseling; inherited diseases; genetics and cancer; and chromosome disorders such as Down syndrome. We also include a chapter on gene therapy, a practice that may hold the key to cures or treatments for many of the disorders we describe in this part of the book.

Part 4: Genetics and Your World

Part 4 explains the broader impact of genetics and covers some hot topics that are often in the news. We explain how various technologies work and highlight both

the possibilities and the perils of each. We delve into population genetics (of both humans, past and present, and endangered animal species), evolution, DNA and forensics, genetically modified plants and animals, and the issue of ethics, which is raised on a daily basis as scientists push the boundaries of the possible with cutting-edge technology.

Part 5: The Part of Tens

In Part 5, you get our lists of ten milestone events and important people that have shaped genetics history, and ten of the next big things in the field.

Icons Used in This Book

All *For Dummies* books use icons to help readers keep track of what's what. Here's a rundown of the icons we use in this book and what they all mean.



HISTORICAL
STUFF

This icon points out stories about the people behind the science and accounts of how discoveries came about.



REMEMBER

This icon flags information that's critical to your understanding or that's particularly important to keep in mind.



TECHNICAL
STUFF

These details are useful but not necessary to know. If you're a student, though, these sections may be especially important to you.



TIP

Points in the text where we provide added insight on how to get a better handle on a concept are found here. We draw on our personal experience for these tips and alert you to other sources of information you can check out.

Beyond This Book

We've included a ton of extra content on the website that accompanies this book. To find it, simply open your favorite web browser, go to www.dummies.com, and search for **Genetics For Dummies** to find the following:

- » **Cheat Sheet:** We've created Cheat Sheet pages that review basic genetics terminology; the structure of cells, chromosomes, and DNA; the laws of inheritance; solving genetics problems; and the basics of transcription and translation.
- » **Bonus chapters:** While the book covers a lot of the hottest topics in genetics, it can't cover everything. Check out the bonus chapter on cloning.
- » **Updates to the book, if any.**

Where to Go from Here

With *Genetics For Dummies*, 3rd Edition, you can start anywhere, in any chapter, and get a handle on what you're interested in right away. We make generous use of cross-references throughout the book to help you get background details that you may have skipped earlier. The table of contents and index can point you to specific topics in a hurry, or you can just start at the beginning and work your way straight through. If you read the book from front to back, you'll get a short course in genetics in the style and order that it's often taught in colleges and universities — Mendel first and DNA second.

1

The Lowdown on Genetics: Just the Basics

IN THIS PART . . .

Discover the basics of genetics and the various careers in the field.

Learn how cells divide and how chromosomes are divvied up among those cells.

Learn about Mendelian genetics and how genes and traits are inherited.

Understand how the inheritance of genes and traits is not always straightforward.

IN THIS CHAPTER

- » Defining the subject of genetics and its various subdivisions
- » A brief introduction to what is covered in this book
- » A review of some of the possible career opportunities in genetics

Chapter **1**

Welcome to Genetics: What's What and Who's Who

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.

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* 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, 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. It wasn't until the 1950s that the importance of the star of the genetics show, DNA, was really understood. 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 (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: 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*, 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 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 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 13, relies on cell biology and an understanding of what happens during cell division. Genetic counseling, which we cover in Chapter 15, also relies on understanding patterns of inheritance to interpret people's medical histories from a genetics perspective. In addition, forensics, covered in Chapter 18, uses Mendelian genetics to determine paternity and to work out who's who with DNA fingerprinting.

Molecular genetics: DNA and the chemistry of genes

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 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 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 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 better treatments, which we address in Chapter 14, focuses on the molecular side of things because tumors result from changes in the DNA, called *mutations*. Chapter 12 covers mutations in detail. Gene therapy, covered in Chapter 16, and genetic engineering, covered in Chapter 19, are subdisciplines of molecular genetics.

Population genetics: Genetics of groups

Much to the chagrin of many undergrads, many aspects of genetics are surprisingly mathematical. 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 17). 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 17. Describing the genetics of populations from a mathematical standpoint is also critical to forensics, as explained in Chapter 18. 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 22).

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.

Genetics as a Career

Studying genetics can lead to a variety of career opportunities, the most common of which we describe in the following sections. The daily life for someone working in genetics can include working in the lab, teaching in the classroom, or interacting with patients and their families. In this section, you'll first discover what a typical genetics lab is like, and then get a quick rundown of a variety of career paths in the ever-expanding field of genetics.

Exploring a genetics lab

A genetics lab is a busy 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, a freezer, and an ultracold freezer 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 that are used to generate more copies of DNA fragments or to determine the sequence of segments of DNA.
- » 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.
- » Computers packed with software for analyzing results and for connecting via the Internet to vast databases packed with genetic information. To get the addresses of some useful websites, see the sidebar, "Great genetics websites to explore," which is located toward the end of this chapter.