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4th Edition

Genetics

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DNA science

Discover the latest advances
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impact your health

René Fester Kratz, PhD
Lisa J. Spock, PhD, CGC

Genetics

for
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4th Edition

**by René Fester Kratz, PhD
Lisa J. Spock, PhD, CGC**

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Genetics For Dummies®, 4th Edition

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Contents at a Glance

Introduction	1
Part 1: The Lowdown on Genetics: Just the Basics	5
CHAPTER 1: Welcome to Genetics: Just About Everyone Loves a Good Pair of Genes	7
CHAPTER 2: Basic Cell Biology	13
CHAPTER 3: Visualize Peas: Discovering the Laws of Inheritance	35
CHAPTER 4: Law Enforcement: Mendel's Laws Applied to Complex Traits	55
Part 2: DNA: The Genetic Material	73
CHAPTER 5: DNA: The Pattern for Life	75
CHAPTER 6: Chromosomes: The Big Picture	91
CHAPTER 7: Replication: A Copy Machine for DNA	105
CHAPTER 8: Transcription: Getting Instructions from DNA	125
CHAPTER 9: Translating the Genetic Code	141
CHAPTER 10: Gene Expression: Finding the Right Tool for the Job	157
Part 3: Genetics and Your Health	173
CHAPTER 11: What Could Go Wrong: Changes in DNA Sequence	175
CHAPTER 12: Chromosome Disorders: It's All a Numbers Game	193
CHAPTER 13: Taking a Closer Look at the Genetics of Cancer	211
CHAPTER 14: Genetic Counseling, Risk Assessment, and Genetic Testing	229
CHAPTER 15: Treating Genetic Disorders and Using Genetics to Tailor Treatment	249
Part 4: Genetics and Your World	263
CHAPTER 16: Tracing Human History and the Future of Populations	265
CHAPTER 17: Solving Mysteries Using DNA	283
CHAPTER 18: DNA Sequencing: Decoding the Genome	305
CHAPTER 19: Genetic Makeovers: Using Genetic Engineering to Change the Genome	319
CHAPTER 20: Giving Ethical Considerations Their Due	339
Part 5: The Part of Tens	353
CHAPTER 21: Ten Hot Issues in Genetics	355
CHAPTER 22: Ten Pathways to a Career in Genetics	365
Index	371

Table of Contents

INTRODUCTION	1
About This Book	1
Foolish Assumptions	3
Icons Used in This Book	3
Beyond This Book	4
Where to Go from Here	4
PART 1: THE LOWDOWN ON GENETICS: JUST THE BASICS	5
CHAPTER 1: Welcome to Genetics: Just About Everyone Loves a Good Pair of Genes	7
Unwrapping Life's Secrets	8
Classical Genetics: Studying How Traits Are Transmitted within Families	9
Molecular Genetics: Diving into DNA Science	10
Population Genetics: Studying the Genetics of Groups	11
Quantitative Genetics: Exploring the Heredity of Complex Traits ...	11
CHAPTER 2: Basic Cell Biology	13
Entering the World of the Cell	14
Cells without a nucleus	14
Cells with a nucleus	16
Exploring Your DNA, Chromosomes, and Genes	18
Examining the basics of chromosomes	18
Finding your genes	21
Mitosis: Splitting Up	22
Step 1: Time to grow	23
Step 2: Dividing up the chromosomes	25
Step 3: The big divide	27
Meiosis: Making Cells for Sexual Reproduction	27
Meiosis I	28
Meiosis II	31
Meiosis in the human body	32
CHAPTER 3: Visualize Peas: Discovering the Laws of Inheritance	35
Gardening with Gregor Mendel	36
Speaking the Language of Inheritance	38
Simplifying Inheritance	40
Establishing dominance	40
Segregating alleles	43

Declaring independence	45
Predicting with Punnetts	45
Finding Unknown Alleles	46
Applying Basic Probability to the Likelihood of Inheritance	47
Solving Genetics Problems.	49
Deciphering a monohybrid cross	49
Tackling a dihybrid cross	50
CHAPTER 4: Law Enforcement: Mendel's Laws	
Applied to Complex Traits	55
Dominant Alleles Rule . . . Sometimes	56
Blending in with incomplete dominance	56
Keeping it fair with codominance	57
Hanging back with incomplete penetrance	58
Alleles Causing Complications	59
More than two alleles	59
Lethal alleles	61
Making Life More Complicated	61
When genes interact	62
Genes in hiding	63
One gene with many phenotypes	64
Genes linked together	65
Uncovering More Exceptions to Mendel's Laws	68
Epigenetics	68
Genomic imprinting	70
Anticipation	71
Environmental effects	71
PART 2: DNA: THE GENETIC MATERIAL	73
CHAPTER 5: DNA: The Pattern for Life	75
Chemical Ingredients of DNA	76
Covering the bases	77
Adding a spoonful of sugar and a little phosphate	79
Assembling the Double Helix: The Structure of DNA	80
Starting with one: Forming the backbone	80
Putting the double in double helix	82
Examining Different Varieties of DNA	85
Nuclear DNA	85
Mitochondrial DNA	86
Chloroplast DNA	87
Digging into the History of DNA	88
Discovering DNA	88
Obeying Chargaff's rules	89
Hard feelings and the helix: Franklin, Wilkins, Watson, and Crick	89

CHAPTER 6: Chromosomes: The Big Picture	91
Packaging the Double Helix	92
Anatomy of a Chromosome	93
Two Chromosomes Are Better than One (or Three)	95
Sex Chromosomes: Is It a Boy or Girl?	97
Sex determination in humans	97
Sex determination in other organisms	102
CHAPTER 7: Replication: A Copy Machine for DNA	105
Unzipped: Creating the Pattern for More DNA	106
How DNA Copies Itself	109
Meeting the replication crew	110
Splitting the helix	113
Priming the process	115
Leading and lagging	116
Joining all the pieces	117
Proofreading replication	118
Replication in Eukaryotes	119
Pulling up short: Telomeres	120
Finishing the job	121
How Circular DNAs Replicate	122
Theta	122
Rolling circle	122
D-loop	123
CHAPTER 8: Transcription: Getting Instructions from DNA	125
RNA: DNA's Close Cousin	126
Using a slightly different sugar	126
Meeting a new base: Uracil	127
Stranded!	128
Transcription: Copying DNA's Message into RNA's Language	130
Getting ready to transcribe	130
Initiation	134
Elongation	136
Termination	136
Post-transcription Processing	136
Adding cap and tail	137
Editing the message	138
CHAPTER 9: Translating the Genetic Code	141
Discovering the Good in a Degenerate	142
Considering the combinations	143
Framed! Reading the code	144
Almost universal	145
Meeting the Translating Team	145

Taking the Translation Trip	146
Initiation	146
Elongation	150
Termination	150
Proteins Are Powerful	152
Recognizing radical groups	153
Giving the protein its shape	154
CHAPTER 10: Gene Expression: Finding the Right Tool for the Job	157
Getting Your Genes Under Control	158
To Be Expressed or Not To Be Expressed?	158
Regulating Eukaryotic Gene Expression: A Time and Place for Everything	160
Controlling Transcription Before It Starts	161
Regulation of Gene Transcription: Flipping the Switch	162
Proteins controlling transcription	162
Sequences controlling genes	163
Hormones controlling transcription	164
Post-transcriptional Control	166
Nip and tuck: RNA splicing	166
Shut up! RNA silencing	167
mRNA expiration dates	168
Gene Control Lost in Translation	169
Modifying where translation occurs	169
Modifying when translation occurs	169
Modifying the protein shape	170
Prokaryotic Gene Expression	170
Bacterial gene organization	171
Bacterial gene expression	171
PART 3: GENETICS AND YOUR HEALTH	173
CHAPTER 11: What Could Go Wrong: Changes in DNA Sequence	175
Heritable or Not Heritable?	176
Facing the Consequences of Sequence Variants	177
Sorting Out Terminology	177
The phenotypic effect of a DNA sequence change	178
The type of DNA sequence change	178
What Causes Sequence Variants?	181
Spontaneous mutation	182
Induced mutations	186
Evaluating Options for DNA Repair	190

CHAPTER 12: Chromosome Disorders: It's All a Numbers Game	193
Chromosome Numbers: No More and No Less	194
Aneuploidy of the Autosomal Chromosomes	196
When chromosomes go missing	196
When too many chromosomes are left in	197
Aneuploidy of the Sex Chromosomes	199
Extra Xs	199
Extra Ys	200
Monosomy X	200
Exploring Variations in Chromosome Structure	201
Duplications	201
Deletions	202
Inversions	203
Translocations	203
Other things that go awry with chromosomes	205
How Chromosomes Are Studied	206
Big enough to see	207
Too small for the naked eye	208
Non-Invasive Prenatal Testing for Aneuploidy	210
CHAPTER 13: Taking a Closer Look at the Genetics of Cancer	211
Defining Cancer	212
Benign growths: Not always so harmless	212
Malignancies: Seriously scary results	213
Metastasis: Cancer on the move	215
Recognizing Cancer as a DNA Disease	215
Exploring the cell cycle and cancer	216
Demystifying chromosome abnormalities	223
Breaking Down the Types of Cancers	224
Hereditary breast cancer	226
Hereditary colorectal cancer	227
CHAPTER 14: Genetic Counseling, Risk Assessment, and Genetic Testing	229
Getting to Know Genetic Counselors	230
Building and Analyzing a Family Tree	231
Autosomal Inheritance: No Differences Among the Sexes	234
Autosomal dominant traits and disorders	234
Autosomal recessive traits and disorders	236
Found on Sex Chromosomes: Sex-linked Inheritance	239
X-linked recessive traits	239
X-linked dominant traits	242
Y-linked traits	243

Sex-limited traits	244
Sex-influenced traits	244
Testing for Genetic Disorders	244
Diagnostic testing	245
Prenatal diagnosis	246
Carrier testing	247
Predictive and susceptibility testing	247
Preimplantation genetic diagnosis	248
Pharmacogenetic testing	248
CHAPTER 15: Treating Genetic Disorders and Using Genetics to Tailor Treatment	249
Alleviating Genetic Disease through Gene Therapy	250
Inserting Healthy Genes into the Picture	250
Finding Vehicles to Get Genes to Work	253
Viruses that join right in	254
Viruses that are a little standoffish	255
Progress on the Gene Therapy Front	255
Utilizing Genetic Information for Precision Medicine	257
Pharmacogenetics (and pharmacogenomics)	257
Cytochrome P450 and drug metabolism	258
Decreasing the risk of side effects of treatment	260
Increasing the effectiveness of treatment	260
PART 4: GENETICS AND YOUR WORLD	263
CHAPTER 16: Tracing Human History and the Future of Populations	265
Genetic Variation Is Everywhere	266
Allele frequencies	267
Genotype frequencies	268
Breaking Down the Hardy-Weinberg Law of Population Genetics	269
Relating alleles to genotypes	270
Populations in balance	271
Violating the law	273
Mapping the Gene Pool	275
One big happy family	276
Ancestry testing	277
Uncovering the secret social lives of animals	278
Changing Forms over Time: The Genetics of Evolution	279
Genetic variation is key	279
Where new species come from	280
Growing the evolutionary tree	281

CHAPTER 17: Solving Mysteries Using DNA	283
Rooting through Your DNA to Find Your Identity.....	284
Investigating the Scene: Where's the DNA?.....	287
Collecting biological evidence.....	287
Moving to the lab.....	289
Employing DNA to Catch Criminals (And Free the Innocent).....	295
Matching the evidence to the bad guy.....	295
Taking a second look at guilty verdicts.....	297
It's All Relative: Finding Family.....	298
Paternity testing.....	298
Relatedness testing.....	301
CHAPTER 18: DNA Sequencing: Decoding the Genome	305
Sequencing: Reading the Language of DNA.....	306
Identifying the players in DNA sequencing.....	306
Finding the message in sequencing results.....	309
Newer, cheaper, faster.....	309
Sequencing Your Way to the Human Genome.....	311
Trying on a Few Genomes.....	315
The yeast genome.....	317
The roundworm genome.....	317
The chicken genome.....	318
CHAPTER 19: Genetic Makeovers: Using Genetic Engineering to Change the Genome	319
Genetically Modified Organisms Are Everywhere.....	320
Old Genes in New Places.....	322
Making a transgenic (recombinant) organism.....	323
Making a transgene using recombinant DNA technology.....	323
Modifying the gene to reside in its new home.....	325
Looking at the GMO Menagerie.....	325
Transgenic animals.....	326
Transgenic insects.....	328
Transgenic bacteria.....	328
Transgenic Plants.....	330
Getting new genes into the plant.....	330
Exploring commercial applications.....	331
Weighing points of contention.....	332
Changing the Blueprint: Gene Editing.....	334
CRISPR-Cas9 gene editing.....	335
Germline versus somatic gene editing.....	337
Discussing the ethics of gene editing.....	337

CHAPTER 20: Giving Ethical Considerations Their Due	339
Profiling Genetic Discrimination	340
Ordering Up Designer Babies	341
Ethical Issues Surrounding Genetic Testing	342
Informed consent	343
Patient autonomy	346
Privacy and confidentiality	346
Incidental findings	347
Direct-to-consumer testing	349
Practicing Safe Genetic Treatments	350
Genetic Property Rights	350
PART 5: THE PART OF TENS	353
CHAPTER 21: Ten Hot Issues in Genetics	355
Direct-to-Consumer Genetic Testing	355
Whole Exome Sequencing	357
Whole Genome Sequencing	358
Stem Cell Research	359
The ENCODE Project	360
Proteomics	361
Gene Chips	362
Evolution of Antibiotic Resistance	363
Circumventing Mother Nature	363
Genetics from Afar	364
CHAPTER 22: Ten Pathways to a Career in Genetics	365
Laboratory Technicians	365
Graduate Student	366
Post-doctoral Fellows	366
Research Scientist	367
College or University Professor	367
Clinical Laboratory Director	368
Clinical Geneticist	368
Genetic Counselor	369
Genetic Counseling Assistant	370
Forensic Geneticist	370
INDEX	371

Introduction

Genetics seeks to understand how the traits of all living things are determined. 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, 4th 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 three 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*, 4th 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.

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 terms 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. The names of genes are also italicized because that is standard practice in the field of genetics.
- » We use **boldface** to indicate keywords in bulleted lists or the action parts of numbered steps.
- » We use monospace for websites and email addresses.

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 traits are inherited. The first chapter introduces you to the field of genetics. 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 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.
- » **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. 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 5, “The Part of Tens”:** In Part 5, you get our lists of ten of the next big things in the field and ten careers in genetics.

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.

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.



WARNING

This icon alerts you to concepts that are typically confusing to people new to the field of genetics. We help you avoid these common misunderstandings.

Beyond This Book

In addition to the abundance of information and guidance related to genetics that we provide in this book, you get access to the following help and information online at Dummies.com:

- » **Cheat Sheet:** To access this book's online Cheat Sheet, go to www.dummies.com and search for "Genetics For Dummies Cheat Sheet."
- » **Bonus chapters:** To access this book's three bonus chapters, go to www.dummies.com/go/geneticsfd4e. You'll find chapters about cloning and defining events in the genetics, as well as a handy online glossary.

Where to Go from Here

With *Genetics For Dummies*, 4th 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.

Explore how cells divide and separate their chromosomes.

Learn about Mendelian genetics and the fundamentals of how genes and traits are inherited.

Go beyond the fundamentals of inheritance to see how multiple genes can interact to determine your traits.

IN THIS CHAPTER

- » Defining the subject of genetics and its various subdivisions
- » Studying the genetics of individuals and families
- » Exploring DNA science through molecular genetics
- » Studying genetic diversity and its evolution within populations of a species
- » Examining how a trait can vary from one individual to another

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.



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.



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 <https://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.)

IN THIS CHAPTER

- » Getting to know the cell
- » Understanding the basics of chromosomes, DNA, and genes
- » Exploring simple cell division
- » Appreciating the complexities of meiosis

Chapter 2

Basic Cell Biology

Genetics is closely related to the study of how cells work. The process of passing genetic material from one generation to the next depends completely on how cells grow and divide. Organisms such as bacteria or yeast that reproduce asexually simply copy their DNA (through a process called *replication*, which is covered later in Chapter 7) and split in two. But organisms that reproduce sexually go through a complicated dance that includes mixing and matching strands of DNA (a process called *recombination*) and then halving the amount of DNA for special sex cells that join to create completely new genetic combinations for their offspring. These amazing processes are part of what makes you unique from every other human on the planet.

In this chapter, we provide a brief introduction to cell structure, DNA, and chromosomes. In addition, you need to be familiar with the processes of *mitosis* (cell division) and *meiosis* (the production of sex cells) to appreciate how genetics works. So come inside your cell and let us introduce you to the basics. Later in this book, we will spend more time on the details of DNA and chromosomes, since these topics lay the groundwork for all things in genetics.

Entering the World of the Cell

There are two basic kinds of cell types, distinguished by whether or not they have a *nucleus* (a compartment filled with DNA surrounded by a membrane):

- » **Prokaryotes:** Organisms whose cells lack a nucleus and therefore have DNA floating loosely in the liquid center of the cell.
- » **Eukaryotes:** Organisms that have a well-defined nucleus to house and protect the DNA.

The basic qualities of prokaryotes and eukaryotes are similar but not identical. Because all living things fall into one of these two groups, understanding the differences and similarities between cell types is important. In this section, you will learn how to distinguish the two kinds of cells from each other, and you will get a quick tour of the insides of cells — both with and without nuclei (plural of nucleus). Figure 2-1 shows you the structure of each type of cell.

Cells without a nucleus

Scientists classify organisms composed of cells without nuclei as *prokaryotes*, which means “before nucleus.” The prokaryotes you’ve probably heard of are bacteria, which are mostly single-celled organisms. Their cells lack nuclei and have relatively small amounts of DNA (you can flip to Chapter 8 for more on the amounts of DNA different organisms possess).

Prokaryotes are the most common forms of life on earth. They exist in every type of environment, including within and on other living things. In fact, at this very moment, trillions of bacterial cells live in and on your body. Most of the bacteria in your body are completely harmless, and others are beneficial to your health; for example, the digestion going on in your intestines is partially powered by bacteria that break down the food you eat. The bacteria that make news headlines, however, are the small percentage that can be deadly, causing rapidly transmitted diseases such as cholera, a severe intestinal disease found most often in populations without a clean supply of water. The vast majority of prokaryotic cells are beneficial to human life and the ecosystems we live in, performing vital functions such as photosynthesis and recycling the dead into usable nutrients for other forms of life. In fact, it’s not too much to say that no other forms of life would survive for long on the planet without the prokaryotes.