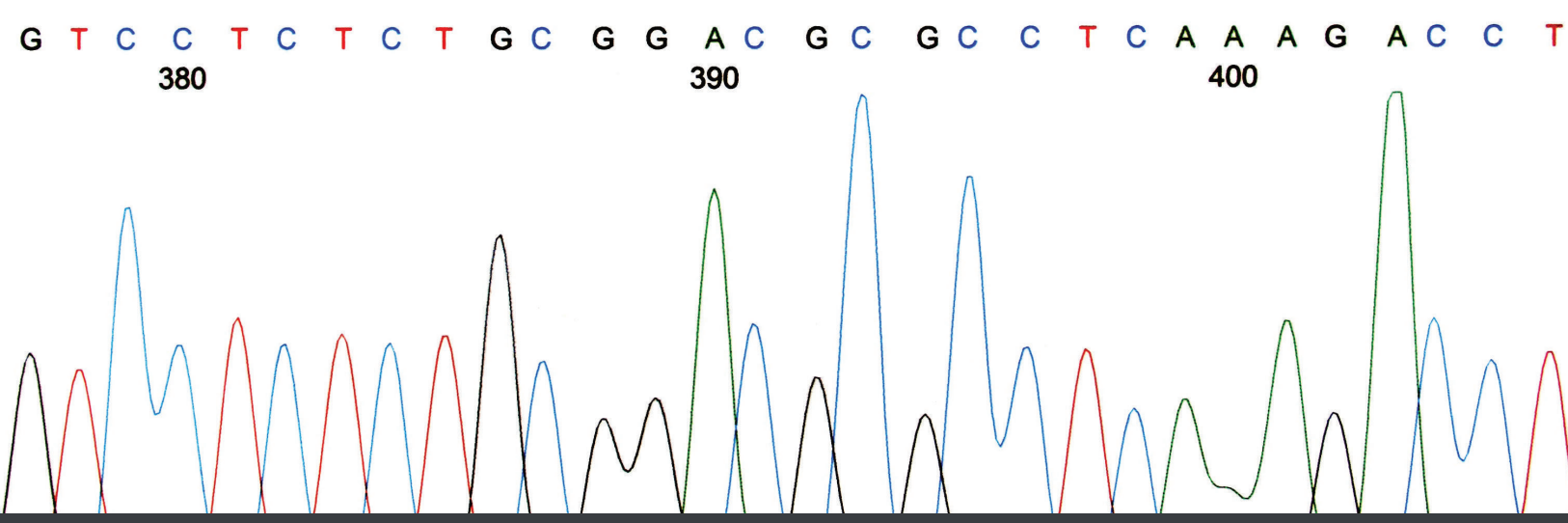

Lab Manual and Workbook for Physical Anthropology

DIANE L. **FRANCE**

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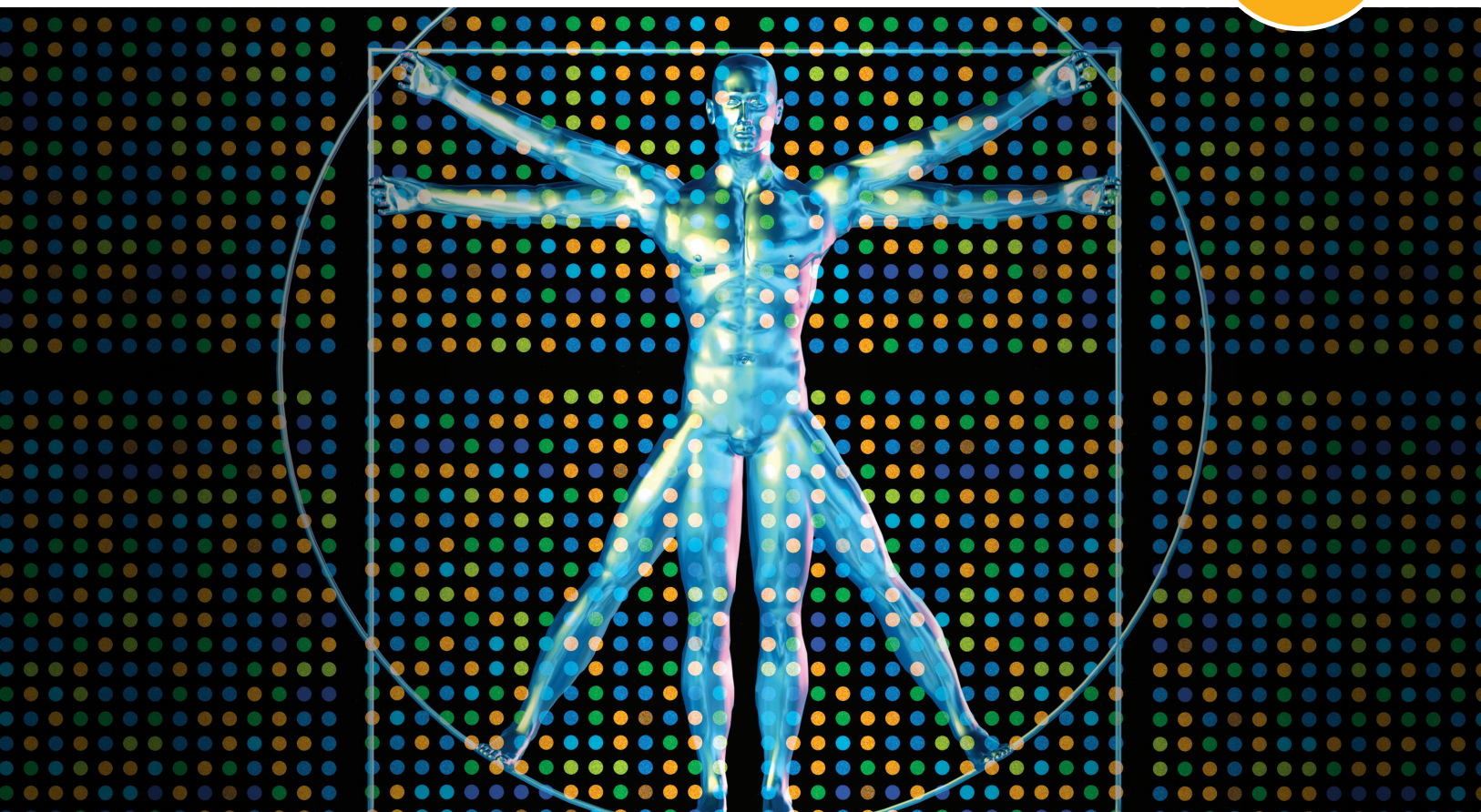

EIGHTH EDITION

Lab Manual and Workbook for Physical Anthropology

Diane L. France

Colorado State University

CENGAGE Learning®

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***Lab Manual and Workbook for Physical Anthropology,* Eighth Edition**
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Library of Congress Control Number: 2016961092

Student Edition:

ISBN: 978-1-305-25904-1

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Printed in the United States of America
Print Number: 01 Print Year: 2017

Brief Contents

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Preface

My goal in writing this text is, and always has been, to provide a foundation of information about physical anthropology and to encourage independent thinking and research. When it is so easy to obtain information (true or not) from the Internet with a few keystrokes, it is more important than ever to have the tools to evaluate whether that information has come from a reliable source, and not just from an opinion page. Education in any discipline and at any level should always stress the importance of critical thinking, and to that end, I expect students to think critically about everything in this book.

The cover of this book reflects the increasing role of genetics in the study of evolution, the relationships of extant primates, and the understanding of humans in general. Current knowledge about the role of genetics in interpreting our world, as well as the research into actually modeling our world through gene therapy and genetically modified organisms (GMOs), is woven through the chapters.

Physical anthropology is a fascinating discipline. It encompasses information about humans and their place in nature more completely than any other field. Within this subject, we explore human biology, how humans have evolved, and how and why we know that we are still evolving. We study our primate cousins in their natural (and unfortunately shrinking) environments, and we compare ourselves to other primate and nonprimate animals. Within the field of physical anthropology, it is possible to explore (and celebrate) the differences and similarities of all modern humans. We examine the human skeleton to discover the wide range of natural variation and learn how to provide clues about the identity of unidentified remains in the medicolegal system. How could anyone not be interested in such a vast and exciting field?

NEW TO THIS EDITION

The eighth edition has been completely updated to reflect recent research. Illustrations and photographs have been improved throughout, and more have been added, and for the first time, the chapters

have been professionally edited. Specific changes made for the eighth edition include the following:

- Real-world controversies and concerns, such as GMOs, the role of gene therapy in medicine, and the presence of Neanderthal genes in modern populations, are present in many chapters.
- Chapters 1 and 2 include up-to-date information about some of the contributions of the Human Genome Project to the understanding of human genetics. After delving into the basics as well as current research, students are asked to evaluate whether society should accept GMOs and whether it is beneficial or even ethical to actually manipulate a person's DNA. Evolution in general, and human evolution in particular, are introduced.
- Chapter 3 still explores human osteology with the addition of side identification and some tips on what bone fragments can tell us about the individual and the bone. More photographs and drawings with labels are particularly useful in those laboratories without adequate osteological material.
- Chapter 4 includes more detail about the growth and development of dentition and bone, with added photographs and updated drawings. The chicken bone exercise gives students the chance to dissect a fried chicken leg, an exercise designed to let them investigate the morphology and mechanics of something very familiar to them, while learning about muscles and bone.
- Chapter 5 explores the theory and practice of species identification and classification and adds the advances in genetics to the concept of species in primates.
- Chapter 6 discusses biomechanical properties of morphological differences in primates and in quadrupedal mammals, leading to later chapters about how primates move and live. This chapter, as well as the following two explore the physical and social characteristics of primates and provide background for the primate fossil record.
- Chapters 7 and 8 still investigate living primate morphology and behavior, with updated photographs.
- Chapters 9 through 12 probe the primate fossil record from before the Paleocene era to more recent prehistory. More illustrations and charts facilitate comparisons between the species, and very recent discoveries have been included. Questions and exercises promote discussion about what these fossil discoveries really mean about how humans and our primate cousins evolved. Recent discoveries of Neanderthal DNA in some modern humans shed a fascinating light on our current variability.

- The remaining chapters further explore modern humans. How do bones respond to disease, genetic conditions, trauma, and intentional modification? How do we use the differences between human populations, age-related changes, and sexual dimorphism to help us determine who an individual was and what happened to that individual at about the time of death? Forensic anthropology is a hot topic today and is covered in some detail in the final chapter.

ACKNOWLEDGMENTS

As with previous editions of this book, many individuals reviewed the seventh edition and suggested changes and additions for the eighth edition. It was apparent that the reviewers thoroughly read the seventh edition, for their suggestions were very helpful. Also, the book has been heavily edited and corrected as per their suggestions.

I also thank the following for generously providing photographs and illustrations: A. Blackstone; Jayne Bellavia; Kathryn Born; Wayne Bryant; the Human Genome Project and developers of genome.gov; M. Y. Iscan; Demris Lee generously provided a valuable section on DNA identification; J. R. Napier; National Museum of Health and Medicine; National Museums of Kenya Casting Program; National Museum of Natural History and the Smithsonian Institution; D. Pilbeam; Plenum Press; W. Sacco; Joseph Henry Press; Shane Walker; D. Ubelaker; Tim White; and Jay Villemarette from Skulls Unlimited provided photographs and access to skeletal materials in his wonderful museum in Oklahoma City.

David Hunt from the Smithsonian Institution was very generous with his time in helping to find subjects for photography for this and previous editions. I also thank Franklin Damann and Brian Spatola from the National Museum of Health and Medicine for providing access to specimens for photography. I also thank them for their help in obtaining what I needed for this book.

I thank all of the people who worked on this book from Cengage Learning and Lumina Datamatics, Inc., including, but not limited to, Elizabeth Beiting-Lipps, Melissa Sacco, Lori Hazzard, Rita Jaramillo, Heather McElwain, and Julia Giannotti.

As always, my husband Art Abplanalp, Jr., was patient and kind through many long days and nights of research, writing, and editing. He is wonderful.

A NOTE ABOUT THE FOSSIL CAST PHOTOGRAPHS

Many of the illustrations of fossil primates and hominins in this book are photographs of casts. The National Museums of Kenya Casting Program, the University of Pennsylvania Casting Program,

the Wenner-Gren Foundation, and the Institute of Human Origins manufactured these casts directly from the original fossils. It is important to understand that these casting programs are assuming all of the risk to the original fossils. It is equally important to understand that a mold made directly from an original specimen will yield the most accurate casts and therefore the most useful casts for instruction.

Money received from the sale of casts produced by the National Museums of Kenya funds research and supports those individuals who are working in the museum. Without that support, the research into our past will suffer greatly. I encourage instructors to purchase casts from those institutions that make molds directly from the originals.

The Scientific Method

OBJECTIVES

1. What is the scientific method?
2. How does the scientific method differ from the belief in intentional causality?
3. Is science just another belief system?
4. How do you set up a scientific experiment?

There are basically two philosophies in understanding the universe and everything in it, although some people combine bits of these philosophies to describe their own belief systems. Those philosophies can be described as "intentional causality" and "natural causality."

Intentional causality (*teleology*) is the belief that everything in the universe is here for a reason and that reason existed before the event or object or individual existed. The modern form of that assertion is contained within the beliefs of intelligent design, which states that the universe and all contained within it are under the intelligent direction of a higher power and that an undirected process such as natural selection cannot explain the final design of living beings or the reason they exist in the first place. In this philosophical model, all change in the world is directed to an end designated by a higher power.

The scientific method (the philosophy followed in this book) does not assume that change is a means to an end or that the end result is designated by a higher power. It is a *process* of testing hypotheses and theories with certain assumptions as a background. Science assumes that there is a *natural* causality and that events, living organisms, and so on, do not appear without something preceding them (also there because of natural causality). Natural causality assumes that change is not directed to a final form or purpose. For every effect there was, and is, a natural (not a supernatural) cause.

Further, there is an assumption in science of *uniformitarianism,* or that the same natural processes operate now as they have in the past, and that the same laws of, for example, physics and chemistry apply everywhere and have for all time. Therefore, hypotheses can be tested because the basic laws that govern them exist everywhere and throughout time.

The scientific method also assumes a high level of *transparency* in reports and publications. This is necessary for the process of testing because if researchers do not know the methods and materials used in an experiment they cannot test the results and conclusions.

EXPERIMENTAL DESIGN: HYPOTHESES AND THEORIES

How many times have you heard "The theory of evolution is just that . . . a *theory*"? This at least implies that it is probably not true, that it is based on false logic, cannot be proven, or perhaps cannot even be adequately described or tested. In fact, a theory is a specific, important step in the scientific method, but it is not the first step.

Before an idea can be said to be a theory, it has to start out as a hypothesis and as a part of the design of an experiment. A hypothesis must be stated in a way in which it can be tested, and it must be stated in such a way that it has the possibility of being proven incorrect. Hypotheses are not stated as a question, but as a statement with an explanation following it (it states what the experimenter thinks will occur). Hypotheses are usually written in the "if–then form" such as "*if* I add fertilizer to one set of geraniums, *then* they will grow bigger than the geraniums to which no fertilizer is added."

A theory is a hypothesis that has been repeatedly tested and the tests have failed to disprove the hypothesis underlying that theory. However, even after a hypothesis has been tested to reach the theory stage, a theory is still testable. A theory is generally recognized as the simplest, most parsimonious explanation (sometimes described as Occam's razor) for a phenomenon, a class of phenomena, or the underlying reality that explains phenomena. It is based on observations that have been tested. These observations are things that you and everyone with normal senses can see or measure with or without aids, and something that can be documented.

If a theory that explains a phenomenon has been observed for a long enough period of time and has not been disproved, it is usually called a "law" or a fact. For example, gravity is explained by the fact (repeatedly tested) that objects with mass attract one another. This is a very simple explanation that can be tested and can be observed. Because this idea has reached the point in scientific testing that it is no longer a hypothesis, it is a theory. In fact, the theories concerning gravity have been tested so many times under so many different controlled conditions that instead of saying that it is the theory of gravity, most people refer to it as the law of gravity. It is generally

recognized as a fact. We all understand that gravity exists and affects our lives in countless ways (from how much we weigh to how much fuel rockets must use to escape the Earth's attraction).

When designing an experiment that will test the hypothesis, it is important to set up a simple hypothesis based on few variables because it can be more easily tested and is potentially falsifiable. (Naturally, if the hypothesis is tested by experimental design and *not* falsified, it does not always mean that the hypothesis is true!) **Independent variables** are the subject of the hypothesis, the experiment, and the variables, and are not controlled. The **dependent variables** are those that can be controlled in the experimental design. Experiments are designed to test cause and effect, so a dependent variable is changed to record the effect of the change on an independent variable. When conducting a scientific study, only one variable at a time should be tested. This is only practical—if you change too many variables in one experiment, you don't know which variable is the explanation for the end result.

Independent variables: The subject of the hypothesis and are not controlled.

Dependent variables: Those that can be controlled in the experiment.

EVOLUTION AND SCIENCE

This book discusses evolution on two levels: evolution on a small scale, as can be observed by everyone in everyday life, and evolution as a series of hypotheses and theories used to describe the changes in organisms (in this book we discuss primates) over time.

Evolution on a small scale is simply a change in gene or allele frequencies from one generation to the next. As we shall discuss in the first two chapters of this book, when organisms multiply through the combination of separate genes from two parents, the offspring have frequencies of alleles that differ from the parents. Farmers take advantage of this when they take a cow that produces a high yield of milk and breed that cow more frequently than a cow that produces less milk. Over a few generations of directed breeding, the farmer's herd as a whole likely produces more milk. This change in gene frequency favoring high milk production over generations is evolutionary change.

The other, larger aspect of evolution discussed in this book concerns how humans and our primate relatives came to be who we are today. This part of evolution contains hypotheses and theories about specifically which fossil led to other fossils. Those theories are based, in large part, on the principle of uniformitarianism (as discussed earlier); that is, the same basic laws in nature that occurred 20 million years ago occur today. Some of the evidence for the theories of evolution include where the fossil was discovered in the world. What other fossils have come from that geologic layer and geographical location? Are there associated floral, faunal, or artifacts that might give clues about its age? What is the morphology of the fossil? How is that morphology similar to modern animals, and what does that tell us about how that animal moved or what it ate?

When researchers at the National Museums of Kenya discover a new fossil, they cannot "test" it by modifying a dependent variable to observe the changes in the independent variable. It is still part of the scientific method, however, because of its transparency and the ability of other researchers to observe the scientific evidence and challenge the conclusions.

Science is a method of critical thinking and testing. To that end, please critically evaluate everything in this book. Do not accept any of this on faith.

The scientific publication should fulfill one of the primary assumptions of the scientific method: *transparency*.

THE SCIENTIFIC PUBLICATION

A few different types of papers are published in scientific journals. Some publications report the results of a scientific experiment, and those papers closely follow the format and content of the experiment (as shown in Table I.1). Other publications may describe an instructive forensic case, an interesting new fossil hominin find, or a new technique in genetic identification, but they usually still describe research related to the topic, the description of the discovery or finding, and the author's conclusions. In these scientific reports, and indeed in any scientific experiment, accurate description is vital! In fact, in many reports and publications, a *diagnosis* of, for example, a pathological condition in a skeleton is less important than a precise *description* of the condition. Why? It comes back to the scientific method—it is important to provide enough information so that other researchers are able to take that information and test it, and perhaps develop an alternative diagnosis.

As stated earlier, the publication that reports on the results of a scientific experiment closely follows the format of the scientific inquiry.

TABLE I.1 Comparisons between the Scientific Method and the Scientific Publication

Scientific Method	Scientific Publication
1. Ask a question.	1. Abstract and Introduction
2. Do background research: Has this question (or one like it) already been researched?	2. Background
3. Construct your hypothesis.	3. State the hypothesis (usually stated in the abstract, but can be stated at this point).
4. Test your hypothesis with an experiment.	4. Materials and Methods
5. Analyze your data and come up with results.	5. State your results.
6. Draw a conclusion about the hypothesis.	6. Draw a conclusion about the hypothesis.
7. Communicate your results and conclusion.	7. Publications and talks communicate your results.

The outline of a scientific publication is as follows:

- **Abstract**: A short synopsis of the question and a brief summary of the results.
- **Introduction:** A basic description of the problem and a statement of the hypothesis.
- **Background:** A history of studies related to the hypothesis and the results of those studies.
- **Materials and Methods:** What did you observe or measure (bones, teeth, fossils, primates, and so on), and how did you measure or observe your subjects?
- **Results**: What were the outcomes of the measurements, observations, statistical tests, and so on?
- **Discussion**: Were you able to falsify your hypothesis? What are your conclusions about the results? In what way(s) would you want to change your research design, materials, methods, and so on, to retest the same or modified hypothesis?
- **References:** List previous work or personal correspondence incorporated in your research.

EXERCISE I.1 NAME _____ SECTION _____ DATE _____

1. Jean-Baptiste Lamarck (1744–1829), a French scientist who believed in evolution, is known for Lamarckism, or inheritance through acquired characteristics. He argued that environmental influences could put pressure on animals, making them use some characteristics more and some less, and that the changes acquired by the parents would be passed to the next generations. A common example of this theory is that giraffes could actually grow longer necks by straining to get to higher and higher branches for food, and that the offspring of these longer-necked giraffes would have longer necks at birth.

   a. Is this argument at the level of a hypothesis or is it a theory? Why?

   b. How would you set up an experiment to test Lamarck's beliefs?

   c. What would be your dependent variables?

   d. What would be your independent variable?

2. Design an experiment to test which company sells the best tomato seeds (the first thing you must do is to define "best"). Be specific when answering the following questions:

   a. What is your hypothesis?

   b. What are your dependent variables?

   c. What is your independent variable?

3. Is anecdotal evidence the same as scientific evidence? Why or why not?

CHAPTER 1

Cellular Genetics

OBJECTIVES

1. What is DNA (deoxyribonucleic acid)?
2. How can we use DNA to show evolutionary relationships between species (discussed in more detail in Chapter 2)?
3. How can DNA be used to solve a crime?
4. Are scientists about to create a genetically engineered human?

The study of genetics is growing and changing at a dizzying speed, particularly after the completion in 2003 of the **Human Genome Project** (HGP), which mapped over 20,000 genes of humans. Scientists are now mapping the genomes of humans in various parts of the world, and are using knowledge about what genes or combinations of genes actually do in the body to research medical advances against disease and genetic conditions. Spin-off projects from the HGP are mapping the DNA of Neanderthals to see how they might be related to modern people (more about that in Chapter 12), and of microbes in the human body to research how they influence our biology and even behavior. To explore this project further, visit http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml.

But before we delve into the incredible advances in genetics, we must first explore the structures of the cell that incorporate DNA into their structure and function.

CELL BIOLOGY

Although DNA is important in all living organisms, we are primarily interested in cells with a nucleus, which include most of the cells of a multicelled (**eukaryotic**) organism. In contrast, single-celled

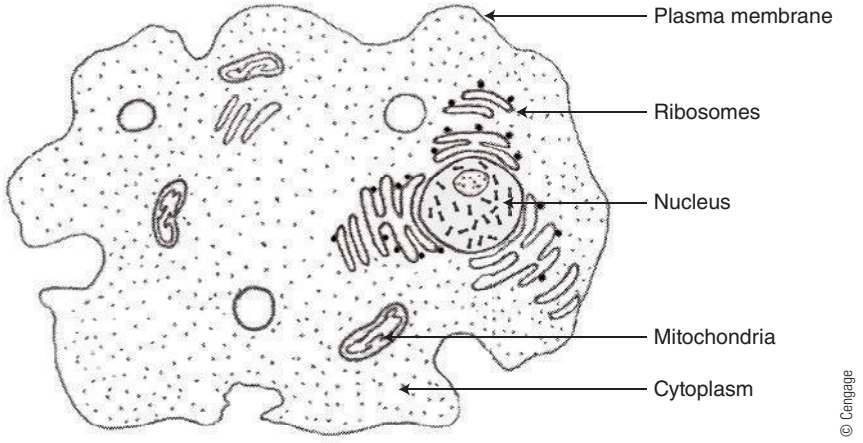


FIGURE 1.1 Structures of a typical cell.

(**prokaryotic**) organisms are those without a discrete nucleus (for example, bacteria). Although we will be primarily interested in structures and events and within the nucleus of the cell, some features of the cell and the organelles (small structures within the cell with specific functions) are also important. These include the following (see Figure 1.1):

- **Plasma membrane:** The outer surface of the cell that protects the cell from the extracellular environment, and that is selectively permeable so as to allow some materials into the cell but keep others out.
- **Cytoplasm:** The fluid within the cell.
- **Mitochondria:** Organelles responsible for the manufacture of energy within the cell. Different cell types have different numbers of mitochondria, depending upon their function. They are located outside of the nucleus, and, interestingly for our discussions, they have their own genome (mitochondrial DNA). As we shall see, nuclear DNA is contributed by the parents to the offspring, but mitochondrial DNA is only contributed by the mother.
- **Ribosomes:** Organelles made up of ribosomal RNA (ribonucleic acid) responsible for the translation of the DNA code within the nucleus into specific proteins. As will be discussed when we explore protein synthesis, **messenger RNA** (mRNA) is transcribed from a section of DNA within the nucleus and it takes that protein code to the ribosomes. **Transfer RNA** (tRNA) recognizes that code and transfers appropriate amino acids to the appropriate location on the mRNA chain.

Messenger RNA: RNA molecules that take information from the DNA within the nucleus to ribosomes outside of the nucleus.

Transfer RNA: RNA molecules that recognize the DNA code on messenger RNA and transfer amino acids to the appropriate location on the mRNA chain.

WITHIN THE NUCLEUS

The nucleus contains **chromosomes**, each of which consists of a series of DNA molecules strung together in long spirals of double strands (called a double helix, which might be visualized as sort of a spiral staircase) (see Figure 1.2a-e). Each strand (or **chromatid**) of DNA is composed of smaller molecules (**nucleotides**), each of which is made of one of four chemical bases: **adenine (A), guanine (G), cytosine (C),** or **thymine (T)** held together with a sugar (deoxyribose) and a phosphate compound (see Figures 1.2e and 1.3). A single strand of DNA is held to the other strand in the double helix by hydrogen bonds (like rungs in a ladder), but note in the diagrams that the molecular shape of the bases are such that adenine can only bind to thymine and cytosine can only bind to guanine. They can be connected in no other way within the nucleus (we will discuss an exception to this rule when the code is carried outside of the nucleus, but within the nucleus, A binds with T and C binds with G).

There are two types of chromosomes in a normal cell: **Autosomes** are the chromosomes that carry DNA, which codes for all genetically determined characteristics except for the determination of the sex of the individual. **Sex chromosomes** (X and Y) determine the sex of the individual (although the presence or absence of only the Y chromosome actually determines sex).

Chromosome number varies by species. Most cells in a fruit fly have 8 chromosomes; there are 40 chromosomes in a house mouse, 24 in a tomato, and so on. Every normal human cell that contains a nucleus (except for sperm and ova, which will be explained later) contains 46 chromosomes, which carry all of the genetic information for that cell and for the organism. These 46 chromosomes consist of 23 pairs; one of each pair is from the mother and one from the father. These are termed **homologous chromosomes**, meaning that they each carry genetic information for the same trait at the same location (although they can carry different information for that trait). DNA codes on chromosomes that carry information for the same trait are called alleles. For example, a particular location (**locus**) on each homologous chromosome codes for the condition of earlobe attachment, but one chromosome may carry the genetic code for attached earlobes, and the other chromosome may carry the code for free earlobes. Also, information for a single trait may be carried on multiple chromosomes. We will discuss this further when we talk about specific genetic traits.

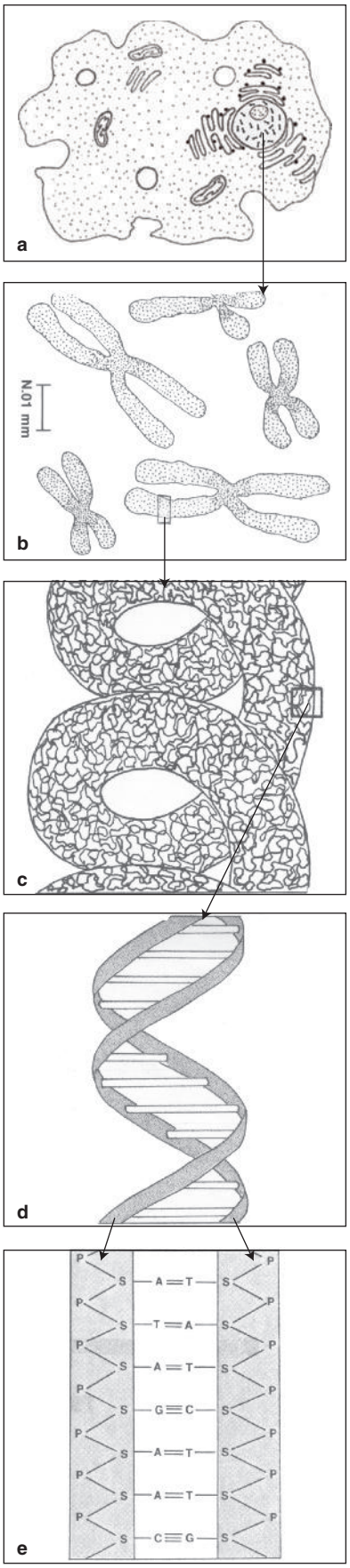


FIGURE 1.2 (a) A generalized cell. Arrow from nucleus: (b) chromosomes; (c) close-up of chromosome; (d) coiled DNA strand; (e) close-up of single DNA strand showing the base, sugar, and phosphate components of a nucleotide.

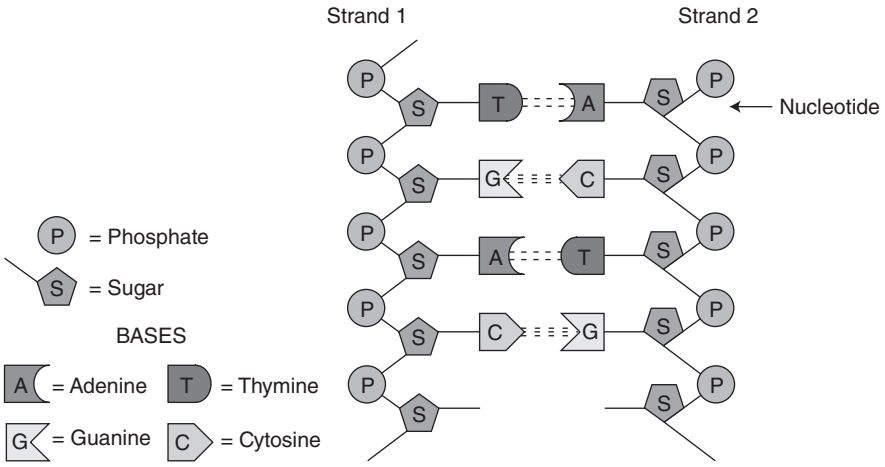


FIGURE 1.3 Nucleotides in a DNA double helix.

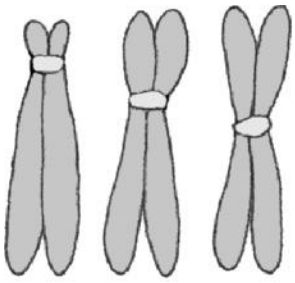


FIGURE 1.4 Positions of a centromere.

Telocentric (left): Centromere at extreme end.

Acrocentric (middle): Centromere off center.

Metacentric (right): Centromere in center.

We can look microscopically at the individual chromosomes and match them with their homologues with a **karyotype** (several sites online allow you to create a karyotype). A karyotype is the summary of the chromosomes within a nucleus and is often viewed as an image with the paired chromosomes lined up according to the position of the **centromere** (the place where two sister chromatids are joined together; see Figure 1.4) and by size (though to truly be able to recognize the homologues so that they may be paired, a dye is often used to create banding patterns within the chromosomes).

Notice on the karyotype shown in Figure 1.5 that there are 22 pairs of autosomes and two sex chromosomes. In this illustration

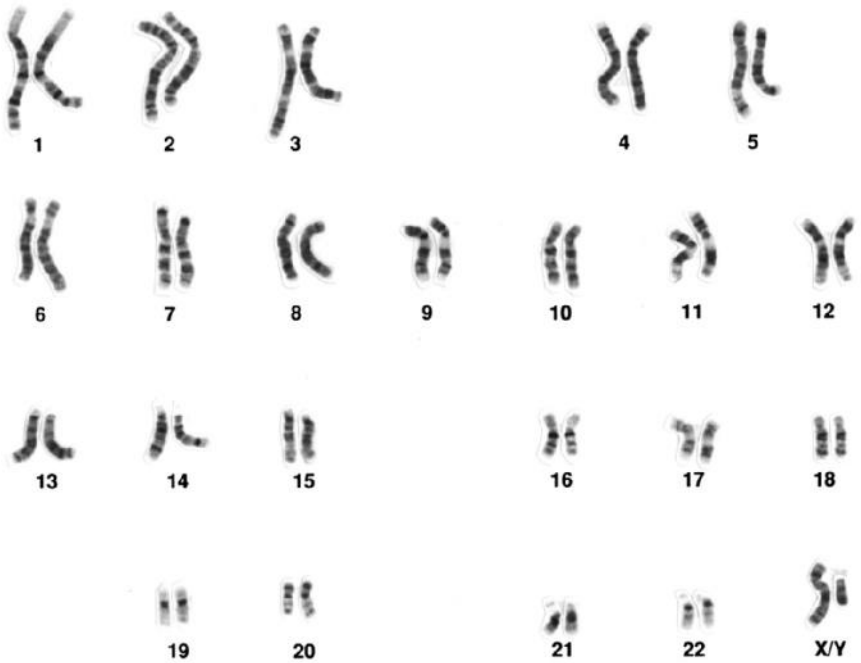


FIGURE 1.5 Normal human male karyotype. *Courtesy: National Human Genome Research Institute*

there is one X and one Y chromosome, so this karyotype is from a normal male. Karyotyping can also assist in demonstrating abnormal genetic sequences, as we will discuss.

NUCLEAR DNA REPLICATION

DNA often has to make copies of itself in whole (during cell replication) or in part (in making proteins outside of the nucleus). The specific processes of cell division and protein synthesis will be discussed later, but this is an appropriate location to explore the basic process common to all DNA replication. When DNA replicates or when a portion of DNA code is needed for protein synthesis, the hydrogen bonds binding the two strands of DNA are broken by an **enzyme** (**polymerase** and other enzymes control different portions of the replication sequence), and the strands essentially "unzip." These hydrogen bonds are shown as double or triple lines in Figure 1.3. The bases (A, T, C, G) on the single DNA strand attract the bases on free nucleotides within the fluid of the nucleus. Those free nucleotides attach themselves to the appropriate loci of the original (now unzipped) DNA chain (see Figure 1.6). Just as in the original DNA chains, adenine can only attach itself to thymine, and cytosine can only attach itself to guanine. Each original single strand of DNA has now paired with a new strand of DNA (these are called **chromatids**), and there are twice as many DNA strands in the nucleus. At this point in humans, there are now copies of 46 chromosomes, or 92 chromatids. The cell can now divide, with each new cell receiving one of the two copies. As will be discussed later, this process is important in many practical applications involving DNA (such as DNA typing).

Enzyme: A protein that accelerates a specific chemical reaction in a living system.

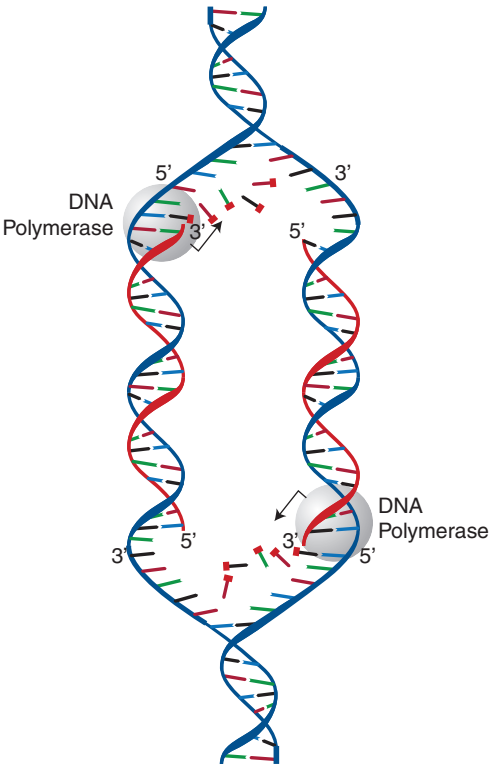


FIGURE 1.6 DNA replication. *Courtesy: National Human Genome Research Institute*

TERMINOLOGY

Allele: Alternative forms of a gene. For example, if the genotype of an individual is Pp for the trait of PTC tasting (see page 38 in Chapter 2), it contains two alleles determining this trait: P and p, one allele for PTC tasting, one for nontasting.

Autosomes: Chromosomes that carry DNA for all characteristics except for the sex of the individual.

Bases: Part of a nucleotide held together with a sugar and a phosphate compound. The bases make up the DNA and RNA chains. DNA bases are adenine, guanine, cytosine, and thymine. RNA bases are adenine, guanine, cytosine, and uracil.

Centromere: Where two sister chromatids are joined.

Chromatid: One of two identical copies of DNA comprising a replicated chromosome.

Codominant alleles: Alleles that, when paired in an organism, are both expressed.

continues

Cytoplasm: The fluid within the cell.

Dominant allele: An allele that is phenotypically expressed in the heterozygote and that prevents the expression of the recessive allele (that is, masks recessive alleles phenotypically). A dominant allele is written in uppercase letters.

Enzyme: Protein that acts as a catalyst for chemical reactions.

Eukaryote: An organism whose cells contain organelles and a nucleus enclosed by a membrane.

Gene: That section of DNA that defines the specific code for a series of amino acids making up a protein or a part of a protein.

Gene pool: The total complement of genes in a population.

Genome: A complete set of chromosomes (and genes).

Genotype: All of the genetic information contained in an organism.

Heterozygote: An organism with unlike members of any given pair or series of alleles at a particular locus. Consequently, this individual produces more than one type of gamete (for example, Aa or Bb).

Homologous chromosomes: Carry genetic information for the same trait at the same location.

Homozygote: An organism whose chromosomes carry identical members of a given pair of alleles. The gametes are, therefore, all alike with respect to this locus. For example, a gamete with the genotypes AA or BB or aa carries only one type of allele for each locus.

Karyotype: The summary of the chromosomes within a nucleus often viewed as an image.

Meiosis: A process of cell replication and division that produces gametes, each with half of the complement of DNA.

Mitochondria: Organelles within the cytoplasm responsible for manufacturing energy within the cell.

Mitosis: Process of cell replication and division in which two "daughter" cells are produced with a full complement of genetic information.

Nucleotide: The molecules that make up strands of nucleic acids like DNA (deoxyribonucleic acid). They are attached to the bases adenine, thymine, cytosine, and guanine (and uracil in RNA).

Phenotype: Characteristic (or combination of characteristics) of an individual visually observed or discernible by other means; for example, tallness in garden peas or color blindness or blood type in humans. Individuals of the same phenotype appear alike but they may have different genotypes.

Plasma membrane: Outer surface of the cell.

Polymerase: An enzyme that assembles long chains of nucleic acids. DNA polymerase assembles strands of DNA.

Prokaryote: A single-celled organism (for example, bacteria).

Recessive allele: An allele that is not expressed when paired with a dominant allele. A recessive allele is written in lowercase letters.

Ribosomes: Organelles made of ribosomal RNA (ribonucleic acid) responsible for protein synthesis.

Sex chromosomes: Determine the sex of the individual.

Trait: A distinguishing characteristic or quality of a phenotype (for example, hair color, blood type, eye color, and so on).

MITOSIS

When a cell divides, the DNA must make full copies of itself so that each new cell receives a full complement of the genetic code.

Mitosis is a process of cell replication and division in which two daughter cells, each with a full complement (46 chromosomes in 23 pairs in humans) of genetic information, are produced from a parent cell with a full complement of genetic information as described in the preceding section on "Nuclear DNA Replication." If there are no mutations, the cells from this division carry genetic information identical to that of the parent cell. Most of the cells in the body replicate throughout life by this process. Because of this process, bones grow in a developing child and skin heals itself after a cut.

We will not be concerned with describing in detail the different phases of mitosis (interphase, prophase, metaphase, anaphase, and telophase) or meiosis in this book, as the individual phases are not as important in this level of discussion as is the knowledge of the outcome.

As cell division proceeds, the chromatids line up along the "equator" of the nucleus, and then separate as cell division occurs. As the cell divides, the chromatids move away from each other, each to a different resultant cell, which now contains 46 chromosomes, still in 23 pairs. For a schematic diagram, see Figure 1.7a.

REMEMBER: In mitosis the cells resulting from cell division carry exactly the same genetic information as the parent cell (as long as there are no mutations).

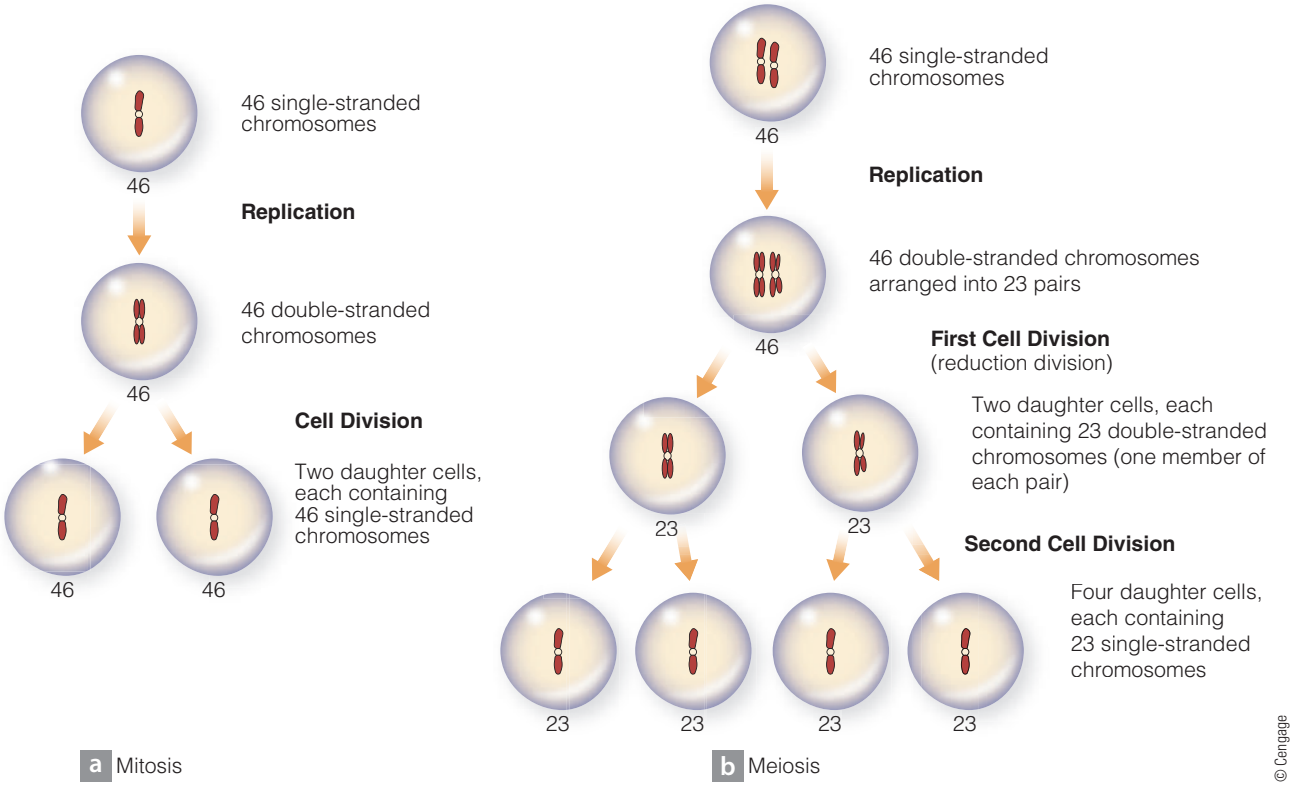


FIGURE 1.7 Mitosis and meiosis. *Source: From Jurmain et al. page 71*

MEIOSIS

Gamete: Sex cells (sperm or ova) that contain half of the number of chromosomes of the adult of that species.

Certain cells participate in the reproduction of the individual by a process of **meiosis,** which creates sex cells, called **gametes** (sperm or ova). Meiosis is the method by which sexually reproducing organisms transfer genetic information through generations; it is therefore important in our discussions of evolution.

The first cell division in meiosis undergoes chromosomal duplication in a manner different from that in mitosis, but it still results in twice the number of chromosomes of a normal cell. After duplication, each set of paired homologous chromosomes (each with two chromatids) lines up along the equator of the nucleus (this is the point in time at which crossing over or recombination can occur, a process that will be discussed in the following section). As the cell undergoes the first division, each member of a paired homologue segregates (separates) into its own daughter cell. After the first division, then, each daughter cell contains one of the homologous chromosomes, but each of these has two chromatids. Each of the daughter cells, in addition, contains only half of the genetic information that had been contained in the parent cell; hence, this first division is also called reduction division.

The two daughter cells next undergo a second mitosis-like division in which the two chromatids of each of the 23 chromosomes separate into two daughter cells. At the end of this division, each of four gametes, or sex cells, contains one copy of one chromosome from the original pair. In other words, each still contains half of the genetic information originally contained within the parent cell, and contains only one copy of each of 23 chromosomes (see Figure 1.7b).

This sequence in meiosis is typical of spermatogenesis, in that for each meiotic cell division, four sperm are formed. During the first division of oogenesis in the female, however, only one viable, functional cell is produced, and that cell in turn produces only one viable, functional cell after the second division. The other nonviable cells are called polar bodies and give up their cytoplasm (future nutrition for the fertilized zygote) to the one viable cell. The end result of oogenesis, then, is one gamete, the egg, which still has only half of the genetic information of the parent cell (as in spermatogenesis), but which is very much larger than a sperm cell. After fertilization, the nucleus of the egg contains half of its genetic information for any trait (except for sex-linked traits) from the father and half from the mother. Each half is in the form of one of the homologous chromosomes.

REMEMBER: In meiosis each daughter cell carries half of the genetic information of the parent cell.

Why must gametes contain only half of the genetic information of a regular body cell? During fertilization of the egg by the sperm, the sperm injects its nuclear genetic information into the egg. If the gametes contained a full complement of DNA, then the resulting cell would contain a “double dose” of DNA, which in most cases is lethal.

CONSIDER THIS

Sometimes chromosomes fail to separate appropriately during meiosis and that causes problems due to an abnormal number of chromosomes in the fetus. This **nondisjunction** can result in only one of a pair of chromosomes (called **monosomy**) or three chromosomes (**trisomy**):

- Trisomy 21 (sometimes called Down syndrome) is caused when the fetus receives three copies of chromosome 21.
- Monosomy X (Turner's syndrome) occurs in females with only one X chromosome. Most females with monosomy X die before birth, but if they survive, they are sterile and usually have skeletal deformities and heart and kidney problems.
- Trisomy X occurs in females with an extra X chromosome, though they often appear relatively normal. Males who inherit an extra X chromosome (Klinefelter's syndrome) sometimes show some breast development and reduced testicular development by puberty. They exhibit reduced fertility and sometimes mental retardation.
- Trisomy 13 (Patau syndrome) have an extra chromosome 13, which causes severe skeletal and organ problems. They have profound mental retardation and usually die at a relatively early age.

EXERCISE 1.1 **NAME** _____ **SECTION** _____ **DATE** _____

Cells and Chromosomes

1. The following sequence of bases is found on one strand of DNA. What is the sequence of bases of the other DNA strand?

   A A C G T T C C G

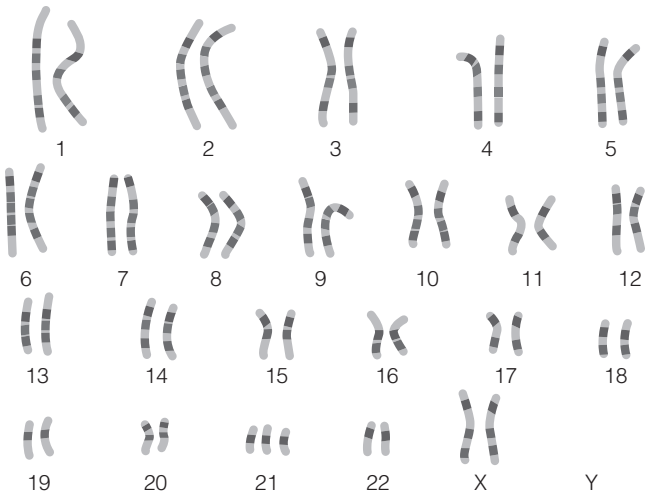


FIGURE 1.8 Karyotype for Question 2. *Courtesy: National Human Genome Research Institute*

2. Figure 1.5 illustrates the karyotype of a normal human male. In Figure 1.8, identify the autosomes and the sex chromosomes.

   a. Is this a male or a female?

   b. This is an abnormal genetic sequence. What is abnormal about it?

   c. Can you diagnose the syndrome?

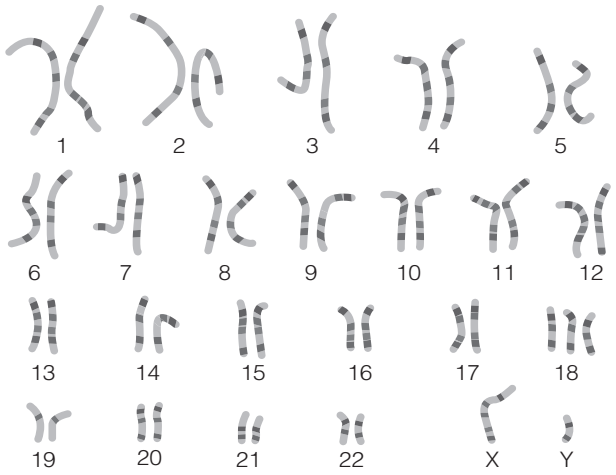


FIGURE 1.9 Karyotype for Question 3.
Courtesy: National Human Genome Research Institute

3. Figure 1.9 is also an abnormal karyotype.

   a. Is this a male or a female?

   b. What is abnormal about this sequence?

   c. Research this abnormality on the Internet and identify the syndrome as well as its consequences.

PROTEIN SYNTHESIS

The DNA in the nucleus of the cell is responsible for more than cell replication in mitosis and gamete formation in meiosis. It is also responsible for providing the blueprint for protein synthesis, which actually occurs outside the cell in small organelles called **ribosomes**. Proteins can be typical structural proteins, enzymes, and hormones, and all are made of smaller molecules called amino acids. These amino acids are defined by three bases, or triplets (remember, the bases are adenine, guanine, cytosine, and thymine). Because this synthesis occurs outside of the nucleus, there has to be a way to get the codes from the nucleus to the ribosomes, and that is the job of the messenger RNA (mRNA) (see Figure 1.10). The nuclear DNA unzips (in a way similar to mitosis though it is called transcription because it "transcribes" the codes), and the bases of the mRNA join with the particular segment of the DNA strand that codes for the prescribed amino acid. The mRNA consists of the bases adenine, guanine, cytosine, and *uracil* (uracil binds with adenine in place of thymine). The strands of mRNA are able to pass through the nuclear wall and translate the "message" from the nuclear DNA to the ribosome. Only a very small percentage of the DNA in the nucleus

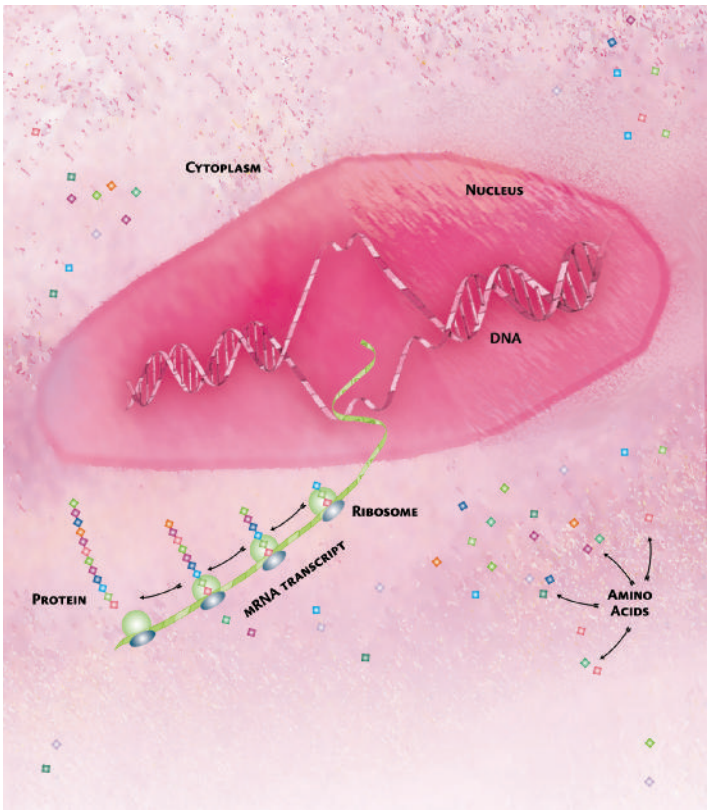


FIGURE 1.10 Transcription. *Courtesy: National Human Genome Research Institute*

Introns: A section of the DNA or RNA molecule that interrupts the code for genes.

codes for proteins or parts of proteins (these areas are called exons in the DNA and in the RNA molecules). There are areas on the DNA strand that do not code for proteins (called **introns**), and if the mRNA strand "reads" an intron, it will snip it out before leaving the nucleus (Strachan and Read 2004). A different RNA—transfer RNA (tRNA)—actually builds the proteins in amino acid blocks. Of the 20 amino acids that exist, 12 are produced by the human body, but 8 (called "essential" amino acids) cannot be manufactured by the body. Those amino acids must be obtained from food (this is why vegetarians must be careful to augment their diet with foods that contain those essential amino acids that would readily be supplied in a diet that contains meat).

Specific proteins are created from specific amino acids, and those amino acids are produced according to the code on the DNA strand (and ultimately on the different RNA strands). The codes for each amino acid consist of three bases (called **triplets** on the DNA chain and **codons** on the mRNA chains); however, for some amino acids, several base triplet codes can code for one amino acid. Likewise, although it has been thought that a specific codon would code for only one amino acid, new research suggests that under certain circumstances a codon may actually be able to code for more than one amino acid (Turanov et al. 2009).

In addition to encoding for proteins, parts of the DNA strand may code for the starting and ending point for coding specific base sequences, and for regulating the expression of other genes elsewhere in the DNA sequence, in other areas of our bodies, or at different times of our lives. Likewise, only a portion of the RNA in a cell is directly involved in the production of proteins (other RNA molecules are involved in diverse functions in the cells). DNA codes and the proteins produced are extremely conservative, such that the basic sequences for protein synthesis and regulatory genes are basically the same from bacteria to humans (Turanov et al. 2009), which is an important consideration in the analysis of evolutionary relationships, as we will discuss in Chapter 2.

SOURCES OF VARIABILITY

Variability can be at the level of the nucleotides and also at the level of the chromosomes. Before and during the first division of meiosis, several events can occur that can alter the genetic configuration of the gametes and, therefore, potentially alter the evolutionary outcome in a population. Most mutations are deleterious or even lethal, but many are neutral or seemingly neutral (that is, they will not immediately alter the selective advantages of the individual). For example, a point mutation occurring during meiosis changes the genetic code in the sperm or egg. If that mutation changes a base to

another base in an area responsible for protein synthesis, and if the resulting code no longer results in a protein required by the body, then that mutation may be lethal or deleterious. But if that mutation changes the base code from ACA to ACG (guanine is substituted for adenine), the resultant amino acid, cysteine, is the same (see Table 1.1, Amino Acids List), so the mutation may have no natural selection advantage or disadvantage. Mutations that appear to be neutral under one set of environmental circumstances, however, may prove to be beneficial or maladaptive under others. Hence, if an environmental change occurs, a neutral mutation is subjected to different selective pressures and may consequently confer advantage or disadvantage to its carrier.

TABLE 1.1 Amino Acids List

DNA Triplets	mRNA Codons	Amino Acid	
ACA, ACG	UGU, UGC	Cysteine	
AAT, AAC	UUA, UUG	Leucine	
AAA, AAG	UUU, UUC	Phenylalanine	Essential
AGA, AGG, AGT, AGC	UCU, UCC, UCA, UCG	Serine	
ACC	UGG	Tryptophan	Essential
ATA, ATG	UAU, UAC	Tyrosine	
CGA, CGG, CGT, CGC	GCU, GCC, GCA, GCG	Alanine	
CTA, CTG	GAU, GAC	Aspartic acid	
CTT, CTC	GAA, GAG	Glutamic acid	
CCA, CCG, CCT, CCC	GGU, GGC, GGA, GGG	Glycine	
CAA, CAG, CAT, CAC	GUU, GUC, GUA, GUG	Valine	Essential
GCA, GCG, GCT, GCC	CGU, CGC, CGA, CGG	Arginine	
GTT, GTC	CAA, CAG	Glutamine	
GTA, GTG	CAU, CAC	Histidine	Essential
GGA, GGG, GGT, GGC	CCU, CCC, CCA, CCG	Proline	
GAA, GAG, GAT, GAC	CUU, CUC, CUA, CUG	Leucine	Essential
TTA, TTG	AAU, AAC	Asparagine	
TAA, TAG, TAT	AUU, AUG, AUA	Isoleucine	Essential
TTT, TTC	AAA, AAG	Lysine	Essential
TAC	AUG	Methionine	Essential (usually a start codon)
TCT, TCC	AGA, AGG	Arginine	
TCA, TCG	AGU, AGC	Serine	
TGA, TGG, TGT, TGC	ACU, ACC, ACA, ACG	Threonine	Essential
ATT, ATC, ACT	UAA, UAG, UGA	Terminating triplets	

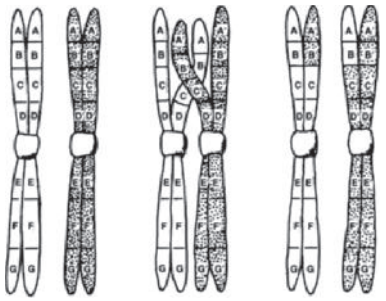


FIGURE 1.11 Recombination at crossover.

Chromosome-Level Variation

During the first division, paired homologous chromosomes may exchange genetic material. This exchange, recombination or crossing over, occurs when the chromosomes are arranged in pairs and after they have duplicated themselves (when there are four DNA strands). Before cell division, the "sister" molecules may exchange sections of DNA (see Figure 1.11). In addition to recombination, chromosomes can be changed by the deletion, duplication, insertion, or other changes of sections of chromosomes (see Figure 1.12). This may alter the specific information carried on a chromosome for a particular trait and can greatly increase the variation of information carried in the gametes. We will talk about recombinant DNA in which sections of DNA are purposely placed in the chromosome in the laboratory shortly.

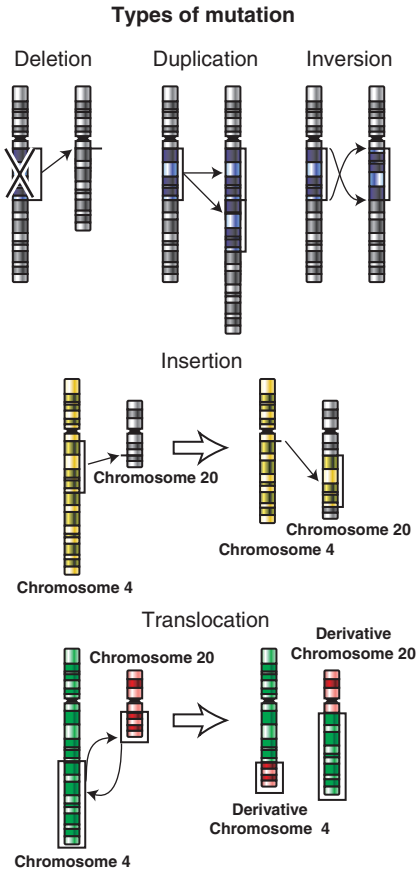


FIGURE 1.12 Types of mutation at the chromosome level. *Courtesy: National Human Genome Research Institute*

Mitochondrial DNA

Mitochondrial DNA (see Figure 1.1) is found outside the nucleus of the cell in its own structures (*mitochondria;* singular: *mitochondrium*). The mitochondrial DNA takes the nutrients we eat and produces energy for the cell. The mitochondrial DNA does not divide during meiosis and is not carried by the sperm to the egg during reproduction (only nuclear DNA is injected into the egg). Because of this, all mitochondrial DNA passed from generation to generation is the DNA from the female (even the mitochondrial DNA in the son's cells), and that mtDNA is an exact copy (barring mutations) from the mother. This form of DNA has become important in discussions about evolution and in the investigation of identity in modern forensic cases. It will also be discussed shortly.

EXERCISE 1.2 **NAME** _____ **SECTION** _____ **DATE** _____

DNA Replication and Protein Synthesis

1. If a point mutation occurred so that adenine was changed to thymine in the amino acid triplet code CTA, what would the resulting amino acid be?

2. If a point mutation occurred so that the second thymine was changed to cytosine in the amino acid triplet code TTT, what would the resulting amino acid be?

3. There are over 17,000 base pairs on chromosome 17 that code for a type of human collagen (COL1A1) found in many tissues, including bone (Bardai et al. 2016; International Human Genome Sequencing Consortium; map can be seen on University of California Santa Cruz genome browser, https://genome.ucsc.edu). If there are significant changes to the normal code for this collagen, it may result in problems with skeletal development and other tissues. Part of the protein sequence for this code is:

   CAGGGCCAGGGGGTCCCTCAGCTCCAGCCTGAGCTCCAGCCTCTCCAT

Fill out the following table containing 10 of those allele codes:

DNA	Complementary DNA Triplet Code	mRNA Code	tRNA Code	Amino Acid
CAG	_____	_____	_____	_____
GGC	_____	_____	_____	_____
GGG	_____	_____	_____	_____
GTC	_____	_____	_____	_____
CCT	_____	_____	_____	_____
CTC	_____	_____	_____	_____
CAG	_____	_____	_____	_____
GAG	_____	_____	_____	_____
CAT	_____	_____	_____	_____
TGC	_____	_____	_____	_____

DNA TESTING AND TYPING

DNA is found in body fluids such as blood, saliva, urine, and semen, soft tissues, bone, teeth, nails, hair roots (nuclear DNA), and hair shafts (mitochondrial DNA; see Figure 1.13). The nuclear human genome consists of approximately three billion base pairs, and, other than identical twins, is unique between individuals. Actually, less than 1 percent of the genome is unique in individuals around the globe, but that is enough to identify any individual with an astounding degree of accuracy. Its uses in forensics are often reported in the news, as it is utilized to identify human remains and trace evidence to perpetrators at crime scenes, although high temperature, humidity, and bacterial activity can degrade the quality of the DNA. DNA has been used in the medical arena for years, tracing mutated genes in families with genetic diseases, and now that the Human Genome Project has mapped the entire human genome, the loci of genes controlling other diseases and genetic conditions continues.

So how does the technology work?

The polymerase chain reaction (PCR), invented by Kary Mullis in 1986, revolutionized DNA analysis. Three main steps occur in PCR analysis: (1) denaturation, in which the two strands of the double helix are separated so that each strand can be used as a template for synthesis of a new strand; (2) annealing, in which primers (short segments of synthetic DNA) bind to the template DNA strands; and (3) extension, in which nucleotides or bases are added to the growing strand of newly synthesized DNA. The three steps are repeated until millions of copies of that section of DNA are created, such that PCR can be used on specimens that were once considered too small or degraded for DNA analysis. A process called "touch DNA" uses

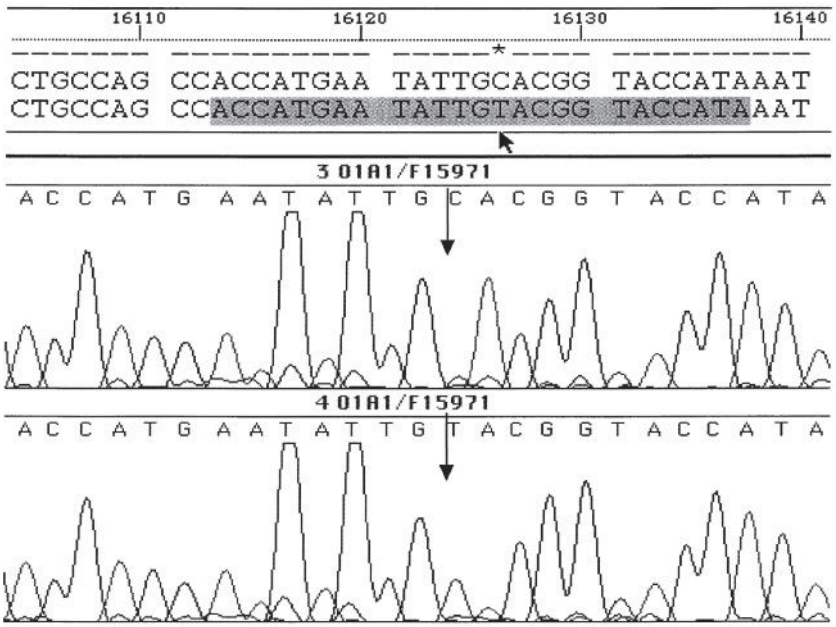


FIGURE 1.13 Mitochondrial DNA sequence analysis. Notice that one allele in the sample (highlighted) is different from the reference sample above it. The reference sample contains a cytosine base at the same location as the thymine base below. *Source: Courtesy of Demris Lee*

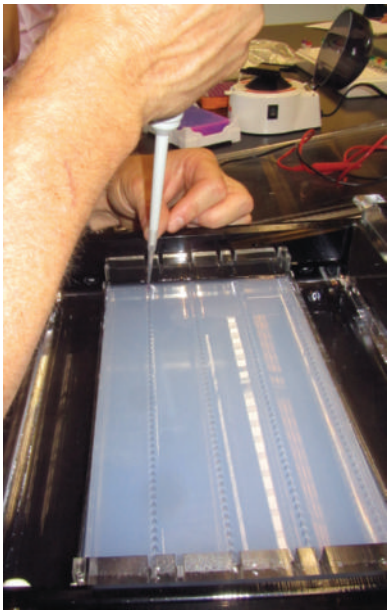


FIGURE 1.14 Adding DNA to the gel strip.

the PCR process so that only a few cells are needed to be able to synthesize enough DNA for typing.

Throughout the human genome, stretches of DNA sequences are repeated over and over. These repetitive sequences are commonly known as variable number of tandem repeats (VNTR). These can be divided into two categories: long tandem repeats (LTRs) or short tandem repeats (STRs). These are distinguished according to the number of base pairs that repeat in a series. LTRs have a large number of repeating base pairs (usually more than seven), while STRs have between three and seven repeating units. If several of these analyses are performed simultaneously for different stretches of DNA sequences, the power to discriminate between individuals can exceed the world's population. See Figures 1.14 and 1.15 for two steps of the process for DNA testing.

In Figure 1.16, a particular DNA sequence of base pairs was repeated three times in Individual 1, four times in Individual 2, and so forth. This segment of DNA is, therefore, of different lengths in these different individuals. When the segment is labeled with a radioactive substance and submitted to electrophoresis (in which the gel is subjected to an electric charge), bands develop on the gel strip. The molecules of DNA will move a distance across that electrically charged gel according to their size (molecules in general will move a distance determined by characteristics other than their size). The largest DNA segment is at the top of the strip, and the shortest at the bottom.

In Figure 1.17, an unknown sample (U) from four different DNA locations is compared with two known individuals. Note that there are two bands for each sample, denoting one DNA segment

The gel used in electrophoresis is either a gelatin made from seaweed, or a compound also used to make soft contact lenses.

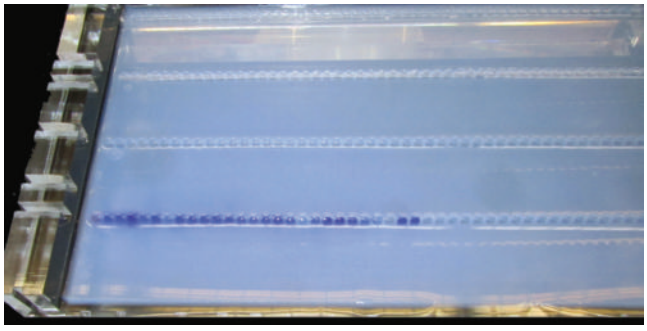


FIGURE 1.15 Electrophoresis starting.

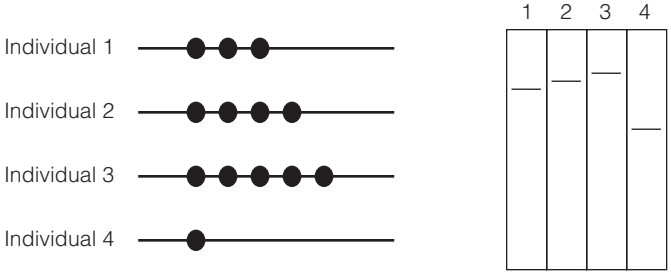


FIGURE 1.16 Variable number of tandem repeats (VNTR) samples from four individuals.

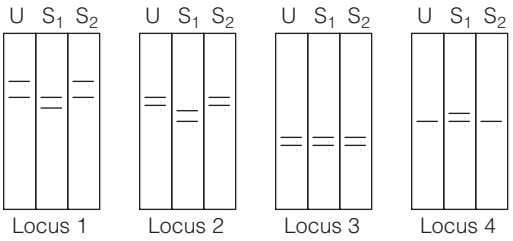


FIGURE 1.17 Unknown "U" compared with two samples.

length from the mother and one from the father. In the event that there is only one band, the segment lengths from the mother and father are the same. In this example, S_2 matches the unknown sample and cannot be excluded from further consideration.

Mitochondrial DNA Typing

The mitochondrial genome is a circular double-stranded molecule consisting of approximately 16,500 base pairs. Most of the differences between individuals are found in the control region, which is the region in the DNA strand that controls the actions of the rest of that region. Instead of two copies, as with nuclear DNA, there are hundreds to thousands of copies of mitochondrial DNA per cell. This is advantageous for typing highly degraded samples (see Figure 1.13 for an example of how a base pair can differ in the sequence of DNA). In fact, even samples as old and degraded as Neanderthal bones have been typed using mitochondrial DNA (more about exciting discoveries in Chapter 12). As was stated previously, mitochondrial DNA is maternally inherited, so assuming mutations have not occurred, all maternal relatives will have the identical mitochondrial DNA sequence. Also, as the mutation rate in mtDNA is greater than nuclear DNA, it can be used to trace inheritance through the female line and can track ancestry for many generations. Mitochondrial DNA is being used to identify the genetic relationship between even distant species.

Similarly, Y chromosomes are only inherited by males in a family. These, too, have the potential to be typed.

The Present and Future of DNA Typing

There is no doubt that DNA typing is a powerful investigative tool, but at present there is a tendency to overstate its power. Note that to use DNA typing, a reference sample must first be obtained. There is no benefit in typing an unknown fragment of tissue or blood stain if there is no person with whom to compare the sample (except to hold the unknown sample until there is a reference). There is at present no mass DNA reference collection, as there is for fingerprints, for example, although since 1994 all states in the United States are required to send DNA samples from many violent criminals after they have been convicted of a crime. The database CODIS (Combined DNA Index System) is searched when unknown DNA is recovered

from a crime scene, and if no match is immediately made, it is stored for future comparisons.

DNA "fingerprinting" is increasingly being used to identify the victims of mass fatality incidents, such as victims of a plane crash; the victims of the 9/11 terrorist acts in Washington, DC, New York City, and Pennsylvania; and victims of other terrorist acts or natural disasters. A decision was made early in the recovery process in New York City after 9/11 to take DNA samples from all remains recovered from "Ground Zero" and to have them analyzed to compare to family members or to objects known to have been used only by the victim (such as a toothbrush, chewing gum, and so on). As was stated previously, we must remember that DNA typing is not useful in all situations (for example, if the remains are badly burned in a plane crash, thereby destroying the DNA). In these situations, family members often demand the "best technology" (that is, DNA typing) for their deceased relatives even when that technology is not practical or possible. In those situations it is sometimes difficult to convince the public that the "low-tech" means of identification such as those discussed in Chapter 15 are the most appropriate.

Significant contributions to the discussion of DNA were made by Demris A. Lee.

CONSIDER THIS

In 1918, after being held captive by the Bolsheviks in the Ipatiev House in Yekaterinburg, Russia, the seven members of the Czar Nicholas II family and four members of his entourage (including his doctor and a maid) were taken to the basement of that farmer's house and killed. In 1991, the Russian government ordered the exhumation of the family members and entourage of the Russian royal family of Czar Nicholas II from a grave outside of Yekaterinburg, where the remains of all but two of the family were finally discovered. The questions before the experts were the following: Are these the remains of the Russian royal family and entourage, and, because two were missing, what members of the family were represented by the remains? No antemortem (before-death) dental records or radiographs (X-rays) were available, so the most promising way to identify the skeletons was through comparison of the DNA, but to whom?

The czar's wife, Czarina Alexandra, was related to Prince Philip, the husband of England's Queen Elizabeth II, in that Prince Philip's maternal grandmother was the czarina's sister. By testing, the researchers discovered that the mitochondrial DNA from the czar's wife and daughters was a match to the DNA from Prince Philip.

Genetically Modified Humans?

We will discuss other genetically modified organisms (GMOs) in Chapter 2 because some of the concern about them involves natural selection, but it is appropriate to start the discussion here when considering modification of genomes.

Consider this example: Over 35 million people around the world are infected with the human immunodeficiency virus (HIV) and many develop acquired immunodeficiency syndrome (AIDS). A gene (CCR_5) that is present in most human beings' genes codes for a protein that allows the virus to attach itself to the body's T cells, part of the body's immune system (actually, more than just this gene plays a part in the HIV story). A very small percentage (less than 1 percent) of the world's population has a mutation that eliminates that gene, and most of them are naturally immune to HIV.

One option for curing the disease is to take stem cells from the bone marrow of a person resistant to the virus and inject them into the infected patient with the hope that the donor's stem cells will repopulate the patient's body with resistant cells. This has actually been successful in one patient, but because patients' immune systems are so compromised by the disease, they can die from fighting the donor's cells before the stem cells have a chance to work. Also, there are so few people with the mutation that this treatment is impractical.

But what if the patient's own stem cells could be genetically modified to eliminate the gene and then inject those cells back into the patient? The patient would not reject his own cells, but you would have to be able to snip out only the offending gene without affecting nearby genes—a very precise target! Various methods are being researched to do just that. Researchers discovered that bacteria have an enzyme (Cas9) that snips out a section of DNA from the genetic code of viruses that try to invade them. Using that enzyme with CRISPR (clustered regularly interspaced short palindromic repeats) technology, it is possible to target only small snippets of DNA and remove them (Kaminski et al. 2016). CRISPR and other technology are being studied to not only remove "malware" in the DNA (or RNA, depending on the situation) but also to insert sections of desirable DNA into the genome of humans, other animals, and plants, and this will be discussed in the next chapter.

At the time of writing this chapter (November 2016), no one has yet claimed to have genetically modified a *viable* human embryo, but Chinese researchers have claimed to modify the genome of a nonviable human embryo, which was destroyed after it was altered (Kang et al. 2016). The attempts to modify the genome for the CCR_5 gene have not always been successful, but the advances in gene modification are fascinating.

EXERCISE 1.3 **NAME** _____ **SECTION** _____ **DATE** _____

DNA Typing

You are asked to analyze a complex case using the evidence provided in Questions 1 through 3.

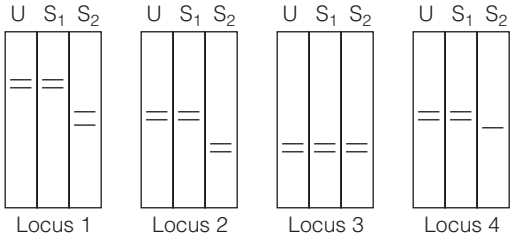


FIGURE 1.18

1. In Figure 1.18, blood samples are taken from a gate (U), an injured woman (S_1), and her former husband (S_2). The woman claimed that she was attacked by her former husband. The man had a cut on his hand, which he explained as a cut from a broken water glass. Is the unknown sample from her wound, from her former husband, or from an unknown third person?

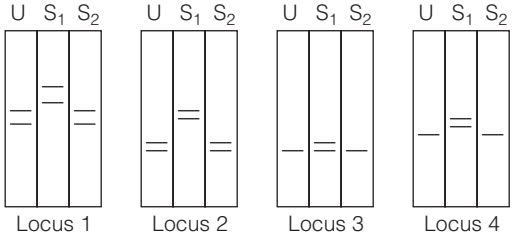


FIGURE 1.19

2. From the same case a second blood sample was collected from a stain on the floor of the former husband's home (see Figure 1.19). Is this stain from the woman, her former husband, or an unknown third person?

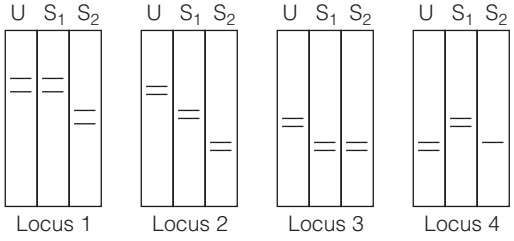


FIGURE 1.20

3. Again from the same case a third blood sample was collected from a knife in the alley two blocks from the woman's home (see Figure 1.20). Is this stain from the woman, her former husband, or an unknown third person?

4. What do these tests tell you about the case?

5. Refer to page 22 about identifying the remains of the Russian royal family. What kind of DNA was used to identify the czar's family's remains? Why was it possible?

6. Would it have been possible to identify which of the czar's daughters was which by using mitochondrial DNA? Why or why not?

TALKING POINTS

- Now that the Human Genome Project has successfully mapped the human genome, there are debates about what to do with the information that now can be obtained about a person's DNA.

  What is your opinion about the following scenarios and why (most of these have actually been suggested by lawmakers and/or insurance companies)?

  - Individuals *convicted* of a felony (homicide, rape, and so on) should have their DNA on file to link them to past and future criminal activity.
  - Individuals *arrested* for a felony should have their DNA on file to link them to past and future criminal activity.
  - Everyone should have their genome mapped:
    - So that health or life insurance companies can analyze the risk of insuring that individual.
    - For identification purposes in mass fatality incidents (plane crashes, and so on).
  - Because the technology is available to perform DNA typing for identification purposes in a mass fatality incident, every element (bodies or body parts, skeletons or skeletal elements and fragments) should be typed, regardless of the cost.
    - Should the families of the victims decide how much money should be spent to identify everyone in a mass fatality incident, or should the medical examiner make that decision? At what point do you draw the line on the amount of money being spent (Thousands? Tens of thousands? Hundreds of thousands?—The costs can easily reach these points in a large disaster).
  - Should we attempt to modify the genome of humans (or other animals or plants)?
    - Under what circumstances should geneticists be allowed to modify a human embryo? How bad should the disease or condition be before embryos should be modified?
    - It is theoretically possible to modify an embryo for sex or even eye color. Should this be permitted?

CHAPTER 2

Population Genetics

OBJECTIVES

1. How does your genetic code express itself in your physical characteristics?
2. How do genetic counselors use these physical characteristics to advise their clients about the genetics of children they may have?
3. How can we determine that evolution is occurring in a population?
4. What are genetically modified organisms (GMOs) and why are some people concerned about them?
5. How can DNA show us the details about evolution?

In this chapter, we will explore genetics at the individual and population levels and discuss how evolution operates. You will explore your own genetic code for certain traits based on how those traits are visibly expressed, and you will learn how to trace the genes and gene frequencies through several generations. We will also discuss the basics of GMOs and explore how GMOs could be beneficial and/or potentially detrimental.

CONSIDER THIS

GREGOR MENDEL

Gregor Mendel was an Augustine monk born in 1822 in Moravia (the southeastern part of the Czech Republic) who was interested in the inheritance patterns of traits in plants and animals. In his time, one of the widely accepted theories of evolution was that offspring inherited characteristics that their parents acquired throughout their lives (championed by Jean-Baptiste Lamarck).

Another theory stated that characteristics of the parents would be blended in the offspring. Mendel's most famous experiments involved pollinating pea plants with, for example, gray and round peas with plants with white and wrinkled peas to determine whether or not the resulting "daughter" peas were a blend of the colors and smoothness of the parent generation. Mendel discovered no blending, but instead, a certain percentage of the offspring had wrinkled peas and a certain percentage had round peas. Through experimentation and mathematical calculations, he developed his principles concerning segregation and independent assortment—the principles we use today and will study in this chapter. Even though his work did not become well known until after his death, he is considered an important figure in modern genetics. For more information, research the many important works about Mendel.

TERMINOLOGY

Allele: Alternative forms of a gene. For example, if the genotype of an individual is Pp for the trait of PTC tasting (see page 38), it contains two alleles determining this trait: P and p, one allele for PTC tasting, and one for nontasting.

Autosomes: Chromosomes that carry DNA for all characteristics except for the sex of the individual.

Chromatid: One of two identical copies of DNA comprising a replicated chromosome.

Codominant alleles: Alleles that are both expressed when paired in an organism.

Dominant allele: An allele that is phenotypically expressed in the heterozygote and that prevents the expression of the recessive allele (that is, masks recessive alleles phenotypically). A dominant allele is written in uppercase letters.

Gene: That section of DNA that defines the specific code for a series of amino acids making up a protein or a part of a protein.

Gene pool: The total complement of genes in a population.

Genome: A complete set of chromosomes (and genes).

Genotype: All of the genetic information contained in an organism; the genetic constitution (gene makeup) of an organism.

Heterozygote: An organism with unlike members of any given pair or series of alleles at a particular locus. Consequently, this individual produces more than one type of gamete (for example, Aa or Bb).

Homologous chromosomes: Carry genetic information for the same trait at the same location.

Homozygote: An organism whose chromosomes carry identical members of a given pair of alleles. The gametes are, therefore, all alike with respect to this locus. For example, a gamete with the genotypes AA or BB or aa carries only one type of allele for each locus.

Phenotype: Characteristic (or combination of characteristics) of an individual visually observed or discernible by other means; for example, tallness in garden peas or color blindness or blood type in humans. Individuals of the same phenotype appear alike but may not have offspring of the same phenotype because the offspring may have different genotypes.

Principle of independent assortment: Paired chromosomes and the alleles on those chromosomes separate into gametes independently of one another. For example, the way in which the

genetic information on chromosome #1 separates into gametes has no influence on how the genetic information on chromosome #3 separates into gametes.

Principle of segregation: During meiosis, paired chromosomes (and therefore alleles) separate into different gametes.

Recessive allele: An allele that is not expressed when paired with a dominant allele. A recessive allele is written in lowercase letters.

Sex chromosomes: Determine the sex of the individual.

Trait: A distinguishing characteristic or quality of a phenotype (that is, hair color, blood type, eye color, and so on).

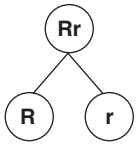
PRINCIPLES OF INHERITANCE

We return to the discussion of gamete production in meiosis to understand how traits are passed from one generation to the next. The key to understanding the principles of inheritance resides in remembering Gregor Mendel's **principle of segregation** and the **principle of independent assortment**. Remember that during meiosis, or the formation of gametes (sex cells), the number of chromosomes in a cell is reduced from 46 to 23 (see Chapter 1). Each pair of chromosomes thus separates, and one chromosome goes to one gamete while the other goes to the other gamete. With the separation of chromosomes, the alleles on those chromosomes also separate.

This is essentially Mendel's principle of segregation, that during meiosis the alleles separate. The principle of independent assortment states that the members of different pairs of alleles assort independently into gametes. Obviously, these principles are true *only* if the alleles in question are on separate chromosomes. Alleles on the same chromosome are not usually independent and, with the exception of some mutations, will stay together during meiosis. As long as a trait isn't lethal or severely deleterious it can "ride along" on the same chromosome with other traits that are beneficial in natural selection. For this reason, it is not valid to suggest that every trait has to have *some* natural selective advantage!

We will assume that all alleles in the following examples and exercises are able to sort independently.

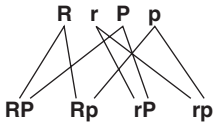
Suppose an individual possessed the two alleles Rr for the trait earwax type (we will explore the specific traits later). In the production of sex cells, these two alleles segregate and go into two different gametes:



These are the gametes, or sex cells.

PTC (phenylthiocarbamide or phenylthiourea) A bitter synthetic chemical that some people (but not all) can taste.

If you wanted to determine the genetics of an individual for two different traits, earwax type and phenylthiocarbamide (PTC) tasting, for example, we could say that his genotype is RrPp. The alleles would separate into gametes as follows:



These are the gametes.

This second example uses the principle of independent assortment; that is, regardless of how the alleles for earwax type segregate, the alleles for PTC tasting will segregate independently, so that every combination possible will occur in different gametes.

CONSIDER THIS

WHY DO WE USE UPPERCASE AND LOWERCASE LETTERS FOR ALLELES?

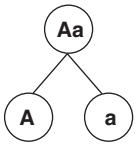
Answer: To keep track of the alleles under discussion, we must follow certain conventions in terminology. Any symbol can be used to designate the alleles, but the letters A, B, C, and so on, or the first letters of the traits being discussed (for example, "P" for PTC tasting) are common. The alleles determining the same trait are written together (PpTt), and the dominant form of the allele is written first. Capital letters designate the dominant form of the allele, and small letters are used for the recessive forms.

SAMPLE PROBLEMS

1. An individual with a genotype Aa would create what kinds of gametes?
2. An individual with a genotype AaBb would create what kinds of gametes?
3. An individual with a genotype AaBbCc would create what kinds of gametes?

Answers:

1. The gametes would be



2. The gametes would be

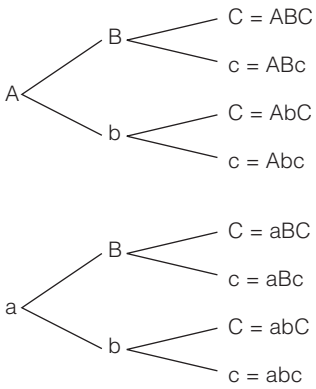
3. The gametes would be

Simple Punnett Square:

	A	a
A	**AA**	**Aa**
a	**Aa**	**aa**

After you have determined the gametes possible for each individual in your mating pair, it is easy to figure out the results of matings between individuals because you simply put the different gametes into all of the possible combinations. One of the easiest ways to do this is by using the Punnett square.

A second method sometimes used to predict the gamete combinations:



Example:

What are the possible outcomes of a mating between individuals AaBb and AaBb? *Hint:* Put the gametes along the top and side of the square and then cross-multiply.

Answer:

The **genotypes** for this cross are:

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

The results in this case are shown here:

AABB (1)
AaBB (2)
AABb (2)
AaBb (4)
AAbb (1)
Aabb (2)
aaBB (1)
aaBb (2)
aabb (1)

There are nine different genotypes for a cross between individuals who are heterozygous for two traits.

What are the **phenotypes** for the preceding cross?

AB (both traits dominant) = 9

Ab = 3

ab (both traits recessive) = 1

Any time you cross two individuals who are heterozygous for two traits, the phenotype ratio will be 9:3:3:1. If you cross two individuals who are heterozygous for three traits, the phenotype ratio will be 27:9:9:9:3:3:3:1 (try it!).

SURVEY OF SOME GENETIC TRAITS

Most phenotypic traits are the results of a combination of effects of heredity and the environment. For example, skin color in an individual is determined to a large extent by genetic and ancestral background but is also affected by the sun and even by diet. A small number of traits have been used traditionally to demonstrate simple Mendelian genetic patterns, although many of these are now known to be affected by much more complex hereditary patterns than once thought. Traits such as tongue rolling and folding (also called "tongue gymnastics"), interlocking fingers and thumbs, and the presence or absence of the palmaris longus tendon are not completely understood but are likely not simply determined by a single allele.

For our exercises, we will assume that the following are inherited as simple Mendelian traits:

1. **Sickle-cell anemia (S, s)** is a condition in which the red blood cells are sickle-shaped (like a "C") instead of disc-shaped. Hemoglobin is a protein molecule in red blood cells that contains iron and transports oxygen and carbon dioxide in the blood. In sickle-cell anemia, the hemoglobin and the shape of the red blood cells are abnormal, which causes problems because the cells cannot deliver sufficient oxygen to the tissues and because the abnormal shape of the cells causes them to get stuck in blood vessels, which causes pain. They can cause a stroke by prohibiting blood flow in areas of the brain, and, in general, the victim suffers from hypoxia (insufficient oxygen). Those individuals with the genotype heterozygous for the sickle trait carry the trait but do not show classic sickle-cell anemia (although under certain conditions, the red blood cells can become sickle-shaped). Those individuals carrying both recessive alleles will show the trait.

   Although sickle-cell anemia is a terrible disease, there is an interesting benefit to being heterozygous for the trait. Malaria is caused by a parasitic organism (*Plasmodium falciparum),*

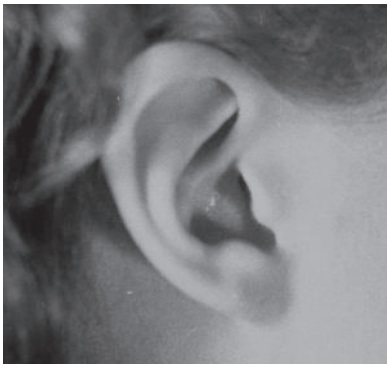


FIGURE 2.1 Attached earlobe (recessive).