Method and Practice in *Biological Anthropology*

A Workbook and Laboratory Manual for Introductory Courses

Second Edition

Samantha M. Hens

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Preface

Welcome! We are pleased to present the long-awaited second edition of *Method & Practice in Biological Anthropology: A Workbook & Laboratory Manual for Introductory Courses*. Since the publication of the first edition in 2006, many other introductory laboratory manuals have appeared on the market offering an array of pedagogical materials. However, choosing a laboratory manual remains difficult for many faculty because while some labs are well equipped with casts and models, others have only a paucity of materials to work with in the classroom. In these latter scenarios, many instructors struggle to provide meaningful lab-based exercises rather than simply repeating lecture materials. This second edition of our popular laboratory manual and workbook is designed to accommodate and function well within various classroom settings.

The 16 chapters in this edition are significantly revised and updated and cover the typical design of introductory courses: genetics, evolution and adaptation, the human skeleton, the nonhuman primates, and our fossil ancestors.

Each chapter has similar pedagogical elements, including:

- A list of chapter objectives
- Essential textual information that can stand alone, or support the formal lecture textbook of your choice
- A set of reading questions to ensure student comprehension of basic concepts and definitions prior to engaging in laboratory activities
  - These reading questions can help to supplement lecture notes when lecture time is limited in the laboratory environment
  - I like to use these reading questions at the start of the class as a pop-quiz; the questions are one way to encourage students to read chapters prior to class and come prepared so that lecture may be minimized and hand-on activities may be maximized!
- An array of laboratory exercises to choose from
  - An assortment of exercises is included to offer instructors a choice of which exercises are best suited to their classroom environment
- A set of post-lab exercises designed to be answered by the student at home
  - These questions build on the information gleaned from the lab work completed in class and provide the students with an opportunity to assimilate the information learned in class and test their understanding by working additional examples on their own
- The design of these chapters allows for assessment of student learning at multiple levels of Bloom's Cognitive Taxonomy, for example, basic definitions/knowledge are learned through reading textual information and answering reading questions; the in-class exercises test a student's comprehension through discussing, demonstrating, and problem solving; and the post-lab questions test the student's recall of class information and ability to apply the concepts on their own.

Teaching an introductory laboratory course is a challenging assignment for any faculty member. It is my hope that this book will also help to make it a rewarding one.

What's New in the Second Edition

With about 8 years between the first and second editions of this text, the changes and updates are numerous throughout and we hope you are pleased with the final product!

Throughout text:

- The former "Pre-Lab" questions have been changed to "Reading Questions" and placed at the front of each chapter, clarifying their role in student learning.
- *We have color images*! New technology in textbook design has made color images an affordable option for students.
- In keeping with modern terminology, humans and our bipedal ancestors are now referred to as hominins, which better reflects genetic and evolutionary relationships, rather than the traditional hominids, which gave preference to human uniqueness over phylogeny.
- *Each and every chapter* has new exercises, additional clarification and explanation of pivotal concepts, and/or expanded areas of coverage.
- *Over 75 new images and figures* are found throughout the text.
- An updated Glossary and Bibliography.

Specifically:

- Chapter 1—the explanation of the nature of science and empirical data has been expanded
- Chapter 3—contains several new Punnett Square examples, emphasizing human traits
- Chapter 4—underwent a major reorganization with a new title; blood typing is now covered before pedigree analysis
- Chapters 7 & 8—have been switched so that the axial skeleton is now presented prior to the appendicular skeleton; extensive skeletal features have been simplified for both chapters with all new images
- Chapter 9—is virtually all new with a new title and greatly expanded sex and age estimation sections and new sections on trauma analysis and pathology
- Chapter 12—contains all new sections on primate community and ecology
- Chapter 13—contains new sections on taxonomy, *Kenyanthropus*, *Australopithecus sediba,* and the Dikika infant along with numerous new images/figures
- Chapter 14—has a new discussion of the Flores, Indonesia find
- Chapter 15—contains an all new section on Neanderthal DNA research and a new section on models for modern human origins
- Chapter 16—is an all new chapter containing anthropometry (originally part of Chapter 9) and new sections on biomedical anthropology and human adaptation

Acknowledgments

Textbooks like this are the product of a team of dedicated people, who support and encourage the author from beginning to end and allow this final product to go to press. I could not have completed this edition without the help of the team at Pearson including my project manager, Bonnie Boehme, editors Nancy Roberts and Carly Czech, photo permissions manager Stephen Merland, and production project manager Jigyasa Bhatia. Thank you to everyone for all of your hard work! Additionally, many thanks to those artists, illustrators, marketing managers, and editors behind the scenes who I may not have personally worked with, but I know you're out there working hard to produce a quality product! Finally, many thanks go to the students who have inspired me over the past 20 years of teaching. This book is for you—to encourage your learning about the place of humans across space and time.

About the Author

Samantha Hens is a professor of anthropology at California State University, Sacramento, with a broad background in teaching and research. Her primary area of expertise is skeletal biology, both human and nonhuman primate. She has worked on a multitude of research projects including age and sex determination from the human skeleton, regression models for stature estimation in paleoanthropological contexts, and geometric morphometrics. The bulk of her work has emphasized three-dimensional modeling of growth and development in orangutan and gorilla crania. More recently, she has studied fluctuating asymmetry in the human cranium using three-dimensional morphometrics in Imperial Roman individuals from central and southern Italy. Currently, she is in the midst of a multi-year project involving improving approaches to age estimation using transition analysis and Bayesian statistics. Dr. Hens regularly teaches courses in Human Osteology, Human Skeletal Analysis, Bioarchaeology, and Paleoanthropology, along with Introduction to Biological Anthropology and Introduction to Forensic Anthropology. In 2006, she was awarded the *Outstanding Teacher Award* in her College. She is a member of the American Association of Physical Anthropologists; the American Association of Anatomists; the Honor's Society, Phi Kappa Phi; and serves on the board for the Omicron chapter of Phi Beta Delta, the Honor's Society for International Scholars. She lives in northern California, where she enjoys yoga and the outdoors.

Chapter 1

The Scientific Method

objectives

After completing this chapter you should be able to:

1. determine the difference between science and other ways of knowing;
2. know the assumptions that all sciences are based upon;
3. describe the basic steps of the scientific method;
4. design a simple experiment to test a hypothesis; and
5. describe the design of a typical scientific research article.

Reading Questions You should read through the text of this lab prior to your class. Then answer the following questions to be ready for the classroom.

1. The idea that the universe is controlled by a supernatural force or deity is best described as a/an:
   a. empiricism,
   b. cause and effect,
   c. teleology,
   d. scientific theory.

2. The first step in the scientific method is:
   a. formulating a hypothesis,
   b. observing an event,
   c. setting up an experiment,
   d. theorizing on the likely result.

3. Scientific reports of experiments are usually reported by the investigators in:
   a. newspapers,
   b. textbooks,
   c. scientific journals,
   d. magazines.

4. The basic assumption in science that all humans experience events in the same way through their senses is called:
   a. uniformity in space and time,
   b. natural causality,
   c. cause and effect,
   d. common perception.

5. A scientific statement that is based on experimental data and has some validity is known as a/an:
   a. conclusion,
   b. theory,
   c. hypothesis,
   d. explanation.

6. The condition or event that may change in an experiment is the:
   a. independent variable,
   b. controlled variable,
   c. original observation,
   d. dependent variable.

7. **True or False:** The results of an experiment do not have to be repeatable.

8. **True or False:** An experiment wherein the researcher cannot control all the variables (which is common in animal behavior studies) is a natural experiment.

9. **True or False:** The variable that researchers try to keep the same for the experimental and control groups is the dependent variable.

10. **True or False:** Evolution is a popular hypothesis in biology, which needs further support to demonstrate its validity.

The Nature of Science

Science is a way of gaining knowledge through critical observation and experiment. It is based on *empirical data*, which is factual information based on observation or experiment. Humans try in many ways to explain their universe, through music, art, literature, religion, philosophy, and science, for example. While many of these approaches may provide answers that we seek, only one, science, is based on empirical tests about nature that can be repeated and verified.

All sciences—including physics, chemistry, molecular biology, geology, and biological anthropology—are based on three assumptions. The first assumption is **natural causality**, which states that all events in nature and the universe are due to natural causes. Thus, scientists assume that everything can be explained by natural means, without reference to a supernatural power or force. Scientific explanations, then, deal with *cause and effect*—the idea that one thing is the result of another, as opposed to *teleological* explanations, which believe that nature has a supernatural design and a purpose. The second assumption is **uniformity in space and time**, which states that all events occur in the same way wherever or whenever they may happen in the universe. Therefore, natural laws, such as gravity, do not change over time or distance. The third assumption is **common perception**, which states that all humans perceive events through their senses in the same way, although morals or ethics may vary. For example, all humans can perceive the color red in the same way through their visual system; however, in some societies red is the color of good luck or fortune, while in other cultures red may symbolize death. Thus, under the idea of common perception, all people, no matter what backgrounds they come from, can come together and speak the language of science to seek knowledge.

EXERCISE 1

Some people claim that epileptic seizures are the result of a supernatural force being directed at a person for punishment of past behavior. Can we test this idea using science? Does it violate any of the assumptions above? If so, which one?

Others claim that epilepsy is the result of neurons misfiring in the brain of afflicted individuals. Can we test this idea using science? Does it violate any of the assumptions above? If so, which one?

The Scientific Method

The **scientific method** is a process for empirically testing possible answers to questions about natural phenomena in ways that may be *repeated* and *verified*. The questions arise from our observations of the world around us. The answers that result from the testing are added to the body of knowledge we have about the natural universe. Scientific explanations are always subject to updates and modifications based on further testing (Figure 1-1). The scientific method generally has the following steps (an example is provided for each step):

1. **Observation.** A researcher may make observations directly from nature with his or her own senses, or from the written words of other investigators who have published scientific articles that are available in university libraries. Either way, the observed phenomenon must be **repeatable**—something that can be observed more than once. These observations typically lead us to question how or why such a thing occurred.
   - *Observation:* Historically, a scientist observed that maggots were found associated with spoiled meat. This led to the question, where did the maggots come from?

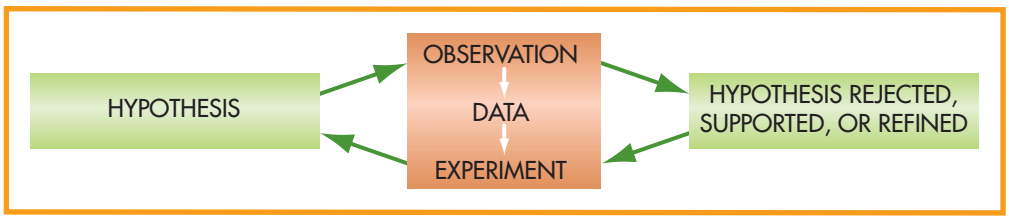


Figure 1-1 The Scientific Method.

2. **Formulate a research hypothesis.** A hypothesis is a proposed explanation for the observed phenomena (i.e., a general answer statement to the question).
   - *Hypothesis:* The maggots arise through spontaneous generation on spoiled meat.
3. **Experiment.** All hypotheses must be *testable.* The testing, or experimental, stage produces more information (data) about the original observation that may or may not support the hypothesis. This stage is often repeated multiple times, by different researchers, who may not have been involved in the original observations.
   - *Experiment:* Spoiled meat was placed into two identical containers, one with no lid and another with a tightly secured lid. Both containers were left next to one another in one room for several days. As the experiment proceeded, flies entered the room and laid eggs on the meat in the opened container. The eggs later hatched into maggots. The meat in the covered container was unaltered.
4. **Form a conclusion.** This may or may not support the original hypothesis. It is a statement based on the data collected and tested in your experiment and has some validity or support.
   - *Conclusion:* The hypothesis was wrong. Maggots do not appear via spontaneous generation, but are the larvae produced by fly eggs.

Sometimes, countless experiments by numerous researchers working in many different countries all support the same conclusion and the results stand the test of time. In these cases, the validity of the results is not seriously doubted by the scientific community, and the conclusion may be considered a **scientific theory**. All scientific theories have been rigorously and exhaustively tested and are supported by a significant body of data. Often the lay public believes that theories are just hunches or not well-supported claims; however, this is a common misconception. Theories continue to be tested and may be modified in the light of new knowledge, especially as we advance technologically. Some examples of modern scientific theories include gravity, the germ theory of disease, heliocentrism, and evolution.

Experimental Design

Experiments test hypotheses. Specifically, they test the relationship between two (or more) variables. Does a change in one variable cause a change in the other variable(s)? In any test, there are three kinds of variables. The **independent variable** is the characteristic of the experiment that is manipulated or changed. The **dependent variable** is the characteristic that is being measured, which may change due to the alterations in the independent variable. The **control variables** are all the other conditions and events, which the researcher attempts to keep the same. Control variables are also independent variables, but they are held constant, whereas the independent variable may be altered in the experiment. It is not always easy to identify all the variables that may affect an experiment. Reading about other people's work is also important because it helps to familiarize us with what has already been done, helps us define the variables, and helps prevent us from repeating the same mistakes others have made.

■ In the preceding maggot example, the dependent variable, or what we are trying to measure, is the presence/absence of maggots; the independent variable that was altered is the open/closed container. The room temperature, humidity, amount of light, and so on are all independent variables that can be controlled or held constant in the experiment.

Consider another example. You are interested to know if a new weight loss supplement assists people in losing weight. You acquire 40 volunteers for a weight loss program to test the supplement. All participants are male between the ages of 20 and 40. They are randomly assigned into two groups of 20 each. Group A takes the supplement, while Group B receives a placebo. The study participants do not know which drug they are taking. All participants eat a similar diet with equal calories each day and get the same amount of exercise. At the end of 90 days, the two groups are compared for the percentage of weight loss. Your hypothesis is that people lose more weight when taking the supplement. The dependent variable—what you are trying to test—is the percentage of weight loss for each group at the end of the study; the independent variable that you manipulated is the supplement/placebo; and the controlled variables are the diet, exercise routines, ages, and sex of the participants. At the end of the study, your conclusion may support or reject your original hypothesis. Either way, several additional questions may present themselves. For example, are there any other variables that can be controlled? How might this study differ if females were tested? Were any of the participants smokers and if so, how might this have affected the results? This is how different scientists will retest the conclusions from previous studies—attempting to examine the question from multiple angles for a more complete picture.

There are two types of experiments:

1. **Controlled experiments.** These occur when an investigator designs an experimental situation, usually in a laboratory. The researcher can alter one variable (the independent variable) and see what effect it has on the other variable (dependent variable). The two examples provided above, maggots and weight loss supplements, are examples of controlled experiments. In all controlled experiments, one group should remain unchanged and unaltered in any way: the *control group* or *comparison group*. This control group is used as a baseline from which to compare the other group(s). The independent variable is not altered in the control group. The control group in the maggot experiment was the covered container and in the weight loss experiment it was the placebo group.
2. **Natural experiments.** The researcher has little or no control over any of the variables in natural experiments. It may be that the researcher cannot control the variables or that it is unethical to do so. Such experiments are common in animal behavior studies, where an investigator spends a considerable amount of time in the field observing subjects and taking careful notes of their observations. This type of experiment is common to some branches of the natural and social sciences.

EXERCISE 2

An experiment is done to test the effect of a new experimental drug for high cholesterol. A group of 200 volunteers are separated into two groups of 100 each. Both groups are instructed to follow a similar diet and activity level. Group 1 is given the experimental drug daily for 90 days, while Group 2 is given a placebo. The individuals in the groups do not know whether they are taking the new drug or the placebo. All participants are tested at the start of the study for their serum cholesterol levels. The average for Group 1 is 310 mg/dL, and the average for Group 2 is 302 mg/dL.

After 90 days, all participants' serum cholesterol is tested with a blood test. The average serum cholesterol level for Group 1 is 299 mg/dL, and the average for Group 2 is 300 mg/dL.

Using this information answer the following:

a. What is the hypothesis being tested? _____

_____

b. What is the dependent variable? _____

_____

c. What are the independent variables? _____

_____

d. Which variables are controlled? _____

_____

e. Which is the control group? _____

_____

f. Did the experiment produce data that supports the hypothesis? _____

_____

EXERCISE 3

An experiment is done to test the effect of a high-fat diet on mice. In all, 50 weanling mice are separated at random into two groups of 25 each. At the start of the experiment all mice weigh approximately the same amount, about 20 g. Group 1 is fed a normal diet with balanced amounts of protein, carbohydrates, vitamin supplements, and fat. Group 2 is fed the same amount of protein, carbohydrates, and vitamin supplements but is given a much higher fat content. The cages are cleaned and mice are given fresh food and water daily.

After 6 months, all mice are weighed. The average weight gain for Group 1 is 8.2 g. The average weight gain for Group 2 is 12.6 g.

Using this information answer the following:

a. What is the hypothesis being tested? _____

_____

b. What is the dependent variable? _____

_____

c. What are the independent variables? _____

_____

d. Which variables are controlled? _____

_____

e. Which is the control group? _____

_____

f. Did the experiment produce data that support the hypothesis? _____

_____

EXERCISE 4

An experiment is done to test the effect of artificial light on geraniums. Seventy-five geranium seedlings are grown in a laboratory. The plants are separated into five groups of 15 plants each. The following table shows the groups, how much light each receives per day, and the average height of the plants after 180 days in the laboratory. All plants are fed the same amount of water and fertilizer daily. Group 4 receives as much light as all the other plants in the laboratory, which are not part of the experiment. Thus 16 hours is average. Groups 1 to 3 receive less light than average, and Group 5 receives more light than average.

Group #	Hours of Light per Day	Height at 180 Days
1	3	5.0 cm
2	6	14.5 cm
3	12	29.2 cm
4	16	36.1 cm
5	24	25.4 cm

Using this information, answer the following:

a. What is the hypothesis being tested? _____

_____

b. What is the dependent variable? _____

_____

c. What are the independent variables? _____

_____

d. Which variables are controlled? _____

_____

e. Which is the control group? _____

_____

f. Did the experiment produce data that supports the hypothesis? _____

_____

EXERCISE 5

A series of observations that might be made by a biological anthropologist are listed at the end of this paragraph. Working in teams, choose one observation from the list, formulate a valid, testable hypothesis, and roughly design an experiment to test your hypothesis. In your work, state your hypothesis, dependent variable, independent variable, and control variable(s).

a. Children from low-income households show evidence of malnutrition.

b. In most humans, the right humerus (upper arm bone) is larger than the left humerus.

c. Expectant mothers who smoke often have low-birth-weight babies.

d. People living on the island of Palau have the highest rates of schizophrenia in the world.

e. Orangutans living in zoos tend to be overweight when compared to their wild counterparts.

The Research Article

The goal of the scientific community is to share information—the results of their experiments—with one another. Scientists are also highly competitive and critically examine and test the work of their colleagues. To this extent, scientists share their work by publishing research articles in scientific journals. Biological anthropologists publish their findings in numerous journals available in the university library and, sometimes, online. Some excellent journals are *The American Journal of Physical Anthropology, The Journal of Human Evolution, Current Anthropology,* and *The American Journal of Primatology.* For a helpful Internet resource that contains articles from both biologists and biological anthropologists go to www.PLOSBiology.org, which is a site sponsored by the Public Library of Science.

EXERCISE 6

a. Review an article from a copy of a typical biological anthropology journal. Then list the titles and functions of the various sections.
   - For example, Abstract—summary of paper
   -
   -
   -
   -
   -

b. In which section do you find the hypothesis being tested, or the study questions?

c. Where would you look to find the details necessary to repeat this experiment?

Science and Religion

As you should now understand, science is based on empirical observation of the natural world. Science's strengths lie in its ability to continually test itself and for people of all different backgrounds and cultures to work for a common explanation. Scientific thinking encourages and requires that individuals question, test, and refine their understanding of the issues. Religious explanations of the world are based on the supernatural and the untestable, usually a spirit or deity. Such explanations provide a complete picture of the world and our place in it, and because this picture is complete, no further observations, hypothesis testing, compiling of data, or revisions of any sort are needed. Religious explanations are usually presented as *dogma*, explanations that are presented in an authoritative manner and are not to be questioned and should be considered absolutely true. Religion is one of the many ways of explaining the world around you, but it violates the assumption of natural causality and does not operate within the scientific method; thus, it cannot be scientific.

Despite these differences, for most people science and religion are compatible in today's world. Science may explain how or when we became human, but it leaves us wanting for answers to such questions as: What is the meaning of life? What does it mean to be a good person? These questions are often answered well by theologians.

Section _____ ID#_____ Name: _____

Post-Lab Questions

1. How does modern science differ from faith? How do these compare in terms of teleological or cause-and-effect explanations? _____

2. Describe the three assumptions all sciences are based on. _____

3. Describe the steps of the scientific method. _____

4. What does an experiment test? _____

5. Describe the differences among the independent variable, dependent variable, and control variable. _____

6. Using either Exercise 2, 3 and/or 4, can you identify any variables that were not controlled for in the study that could have been controlled? _____

7. Reviewing your team's answers to Exercise 5, can you think of an alternate test of your hypothesis? Was anything left out of your original experiment? _____

_____

_____

_____

8. Choose a research article from a physical anthropology journal online or in the library. Can you identify what hypothesis the author(s) is/are testing? Is the experiment designed in such a way that it might be repeated by another investigator? _____

_____

_____

_____

Chapter 2

Cell Biology and DNA

objectives

After completing this chapter you should be able to:

1. describe and label the parts of the eukaryotic cell;
2. describe the types of cells found in the human body;
3. describe chromosome structure and identify human karyotypes;
4. describe the differences between mitotic and meiotic cell division;
5. understand the importance of crossing over and recombination;
6. describe DNA structure, DNA replication, transcription, and translation; and
7. calculate the sequence of bases in DNA or RNA when provided with the complementary strand, and translate the codons into amino acids using the chart.

Reading Questions You should read through the text of this lab prior to your class. Then answer the following questions to be ready for the classroom.

1. Which of the following individuals was responsible for coining the term *cell*?
   a. Hooke,
   b. Darwin,
   c. Wilkins,
   d. Watson.

2. Prokaryotic cells are distinguishable from eukaryotic cells because prokaryotes do *not* contain:
   a. organelles,
   b. a plasma membrane,
   c. DNA,
   d. a nucleus.

3. Chromosome strands are called:
   a. centromeres,
   b. alleles,
   c. chromatids,
   d. homologues.

4. Alternate forms of a gene are called:
   a. alleles,
   b. sister chromatids,
   c. homologues,
   d. replicated DNA.

5. Sister chromatids separate during nuclear division in:
   a. mitosis,
   b. meiosis I,
   c. meiosis II,
   d. both a and c.

6. Who won the Nobel Prize in 1962 for identifying the structure of DNA?
   a. Hooke,
   b. Meischer,
   c. Watson and Franklin,
   d. Watson, Crick, and Franklin.

7. Which of the following is a possible base pairing in DNA?
   a. adenine-cytosine,
   b. adenine-thymine,
   c. cytosine-thymine,
   d. thymine-guanine.

8. Transcription in DNA:
   a. results in the formation of an identical DNA strand,
   b. results in the formation of mRNA,
   c. happens in the nucleus,
   d. requires the assistance of tRNA anticodons.

9. **True or False:** DNA replication occurs in the ribosome.

10. **True or False:** Crossing over is an important source of variability.

Cells and Cell Structure

The **cell** is structurally and functionally the basic unit of life for all organisms. The term *cell* comes from Robert Hooke, who in the seventeenth century observed that cork was made up of small units, which reminded him of the "cells" or cubicles in which monks lived. All cells must arise from preexisting cells, and living organisms may be single celled or multicellular in composition.

Cells may be classified into two types, depending on the presence of an internal, membrane-bound **nucleus**: (1) **prokaryotic** cells, which do not have a separate nucleus and are found in bacteria and cyanobacteria and (2) **eukaryotic** cells, which contain a true nucleus and make up all other forms of life. It is thought that the prokaryotic cells evolved first and that eukaryotic cells evolved from these simpler forms. All eukaryotic cells share certain structural features in common, including (1) a *plasma membrane,* separating the contents of the cell from the outside world; (2) *cytoplasm,* a gel- or fluidlike matrix within the plasma membrane; (3) *organelles,* the various structures responsible for cell functions, such as metabolism and protein synthesis; and

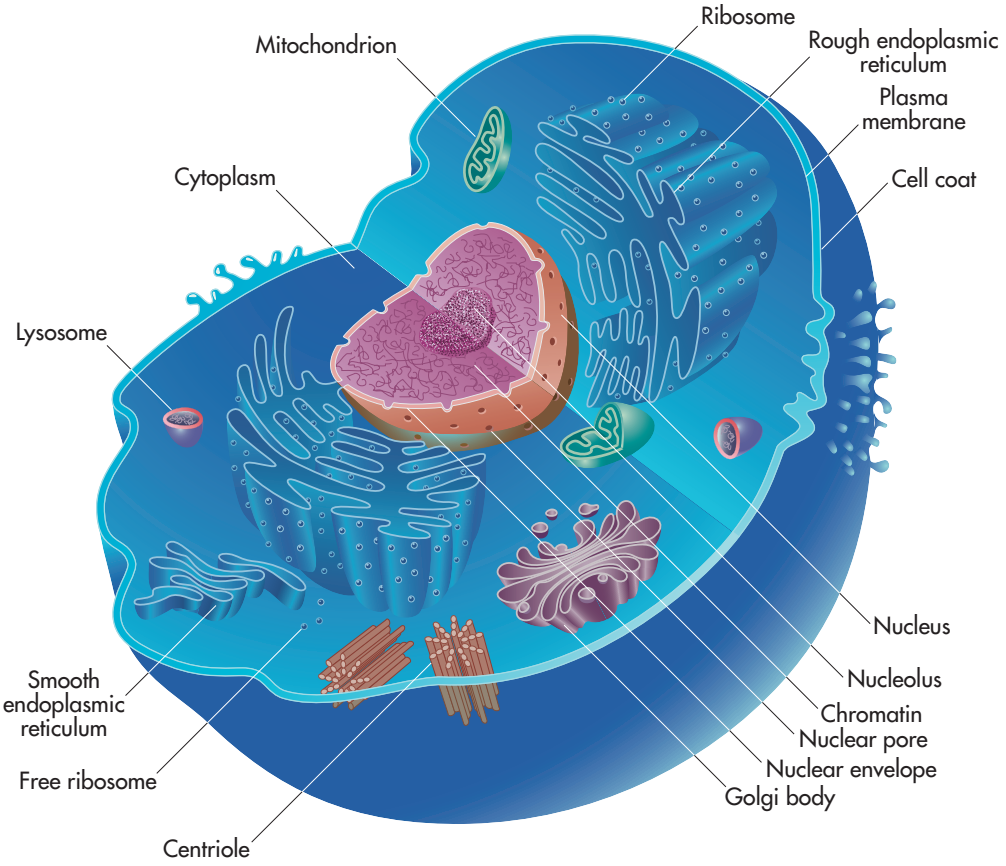


Figure 2-1 A typical eukaryotic cell.

(4) genetic information, in the form of **DNA** (deoxyribonucleic acid), which is stored in the nucleus (Figure 2-1).

Two types of cells are found in humans: (1) **somatic cells** and (2) **gametes**. The somatic cells are those cells that make up the body of an organism—everything from hair and skin to lungs, liver, muscles, blood, and bone. Somatic cells are often referred to as *body cells*. In contrast, the gametes, or sex cells, are the sperm found in the male testes and the ovum (plural ova, or egg cells) found in the female ovaries. The gametes carry the genetic information required to make the next generation.

Chromosome Structure

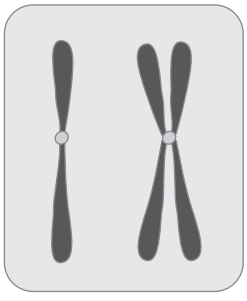


Figure 2-2 Single-stranded and double-stranded chromosomes.

DNA and proteins are found on **chromosomes**. Chromosomes are located in the nucleus and are made up of long, threadlike material called *chromatin* that coils and condenses when a cell is about to divide, making the chromosome visible with a light microscope. Chromosomes are normally single stranded, but they become double stranded when DNA replicates itself, prior to cell division. Each strand is called a **chromatid**. When the chromosome is double stranded, the two chromatids are called *sister chromatids*. A constricted area, called the **centromere**, separates one chromatid strand into two "arms." Chromosomes come in different sizes and can be identified by size and position of their centromere. Centromeres may be in the middle of the chromosome, so that the arms are of approximately equal length, or they may be off center, making the arms of unequal length (Figure 2-2).

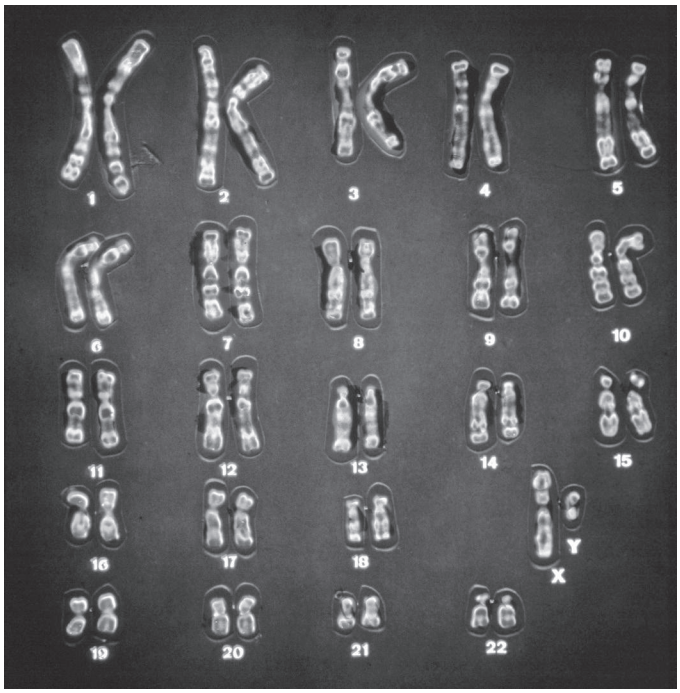


Figure 2-3 A human karyotype. Notice the autosomes, sex chromosomes, and basic chromosome structure.

Different species have a different number of chromosomes in their cells. Normal human body cells contain 23 pairs of chromosomes (46 chromosomes total). The chromosomes are numbered 1 through 23. In other words, there are two copies of number 1, two copies of number 2, and so on, until you reach the two copies of number 23. These are called **homologous pairs**. The members of each pair are similar in size, position of the centromere, and genetic information carried (always for the same traits) (Figure 2-3). This genetic information is distributed along the length of the chromosome as **genes**, segments of DNA that code for specific traits. **Alleles** are alternate forms of a gene; for example, the gene for eye color may have several alleles, such as brown, blue, and green. The first 22 pairs of chromosomes are called the *autosomes.* The final, 23rd pair, are called the *sex chromosomes.* Human females have two X chromosomes as their 23rd pair, while human males have one X and one Y chromosome as their 23rd pair (because these two chromosomes are different, they are not homologous). Human sex cells, or gametes, carry only one copy of each pair, so they have 23 chromosomes in each sperm or ovum.

EXERCISE 1

Can you think of any reason why the gametes have only 23 chromosomes, one of each pair? _____

_____

_____

_____

Chimpanzees have 48 chromosomes in their somatic cells. How many chromosomes do you think are found in their sex cells? _____

EXERCISE 2

When chromosomes are stained and photographed, the resulting image is called a **karyotype**. Chromosomes in a karyotype can be cut out and lined up. They may be arranged into pairs by matching their size and position of the centromere. The position of the centromere may be described as follows:

- **Acrocentric:** At one end, so arms are of unequal length
- **Metacentric:** In the middle, so that arms are of similar length
- **Telocentric:** All the way at one end, so that one arm is barely visible

Sometimes, an individual is born with the wrong number of chromosomes. This is caused by a **nondisjunction**, a failure of the chromosomes to segregate properly during cell division. In this case, you may have a different number of chromosomes in your karyotype due to a deletion or duplication of a chromosome. Some common abnormalities are:

- **Turner syndrome:** X0, deletion of a sex chromosome; these are females who tend to be shorter than average, with below-average intelligence, and are infertile.
- **Klinefelter syndrome:** XXY, an extra X chromosome; these males are taller than average, with below-average intelligence, and are infertile.
- **XYY:** An extra Y chromosome; these are males who may show a tendency toward aggressive behavior.
- **Down syndrome:** Three copies of chromosome 21 (trisomy 21); these individuals are characterized by a suite of traits, primarily mental retardation.

Working individually or in pairs, you will be provided a copy of a standard human karyotype form and a human karyotype. Carefully, cut out the chromosomes and matching pairs, then glue or tape the chromosomes onto the form. When you have finished, answer the following questions:

a. Is the individual whose karyotype you have a male or a female? How do you know? _____

b. Draw a circle around the sex chromosomes in your karyotype and label them. Do the same for the autosomes.

c. Are there any anomalies in the karyotype you have completed? If so, describe it. Be sure to highlight it and indicate the name of the syndrome on your karyotype. _____

Cell Division

Cell division in eukaryotes requires two processes: division of the cytoplasm (*cytokinesis*) and division of the nucleus, **mitosis** or **meiosis**. Cytokinesis ensures that each end product (*daughter cell*) receives the cellular structures needed for life, such as cytoplasm and organelles. Mitosis and meiosis are different types of divisions and result in different numbers of daughter cells. Figure 2-4 provides an illustration of the two processes.

Mitosis. *Fertilization* is the fusion of egg and sperm nuclei, resulting in a single-celled **zygote**, or fertilized egg. This zygote then divides by mitosis into two cells, which in turn divide into four, and so on. Eventually a multicellular organism is produced. During each cell division, each daughter cell receives a complete set of genetic information and the necessary cellular components. This cell division occurs during growth and later repair of body tissues.

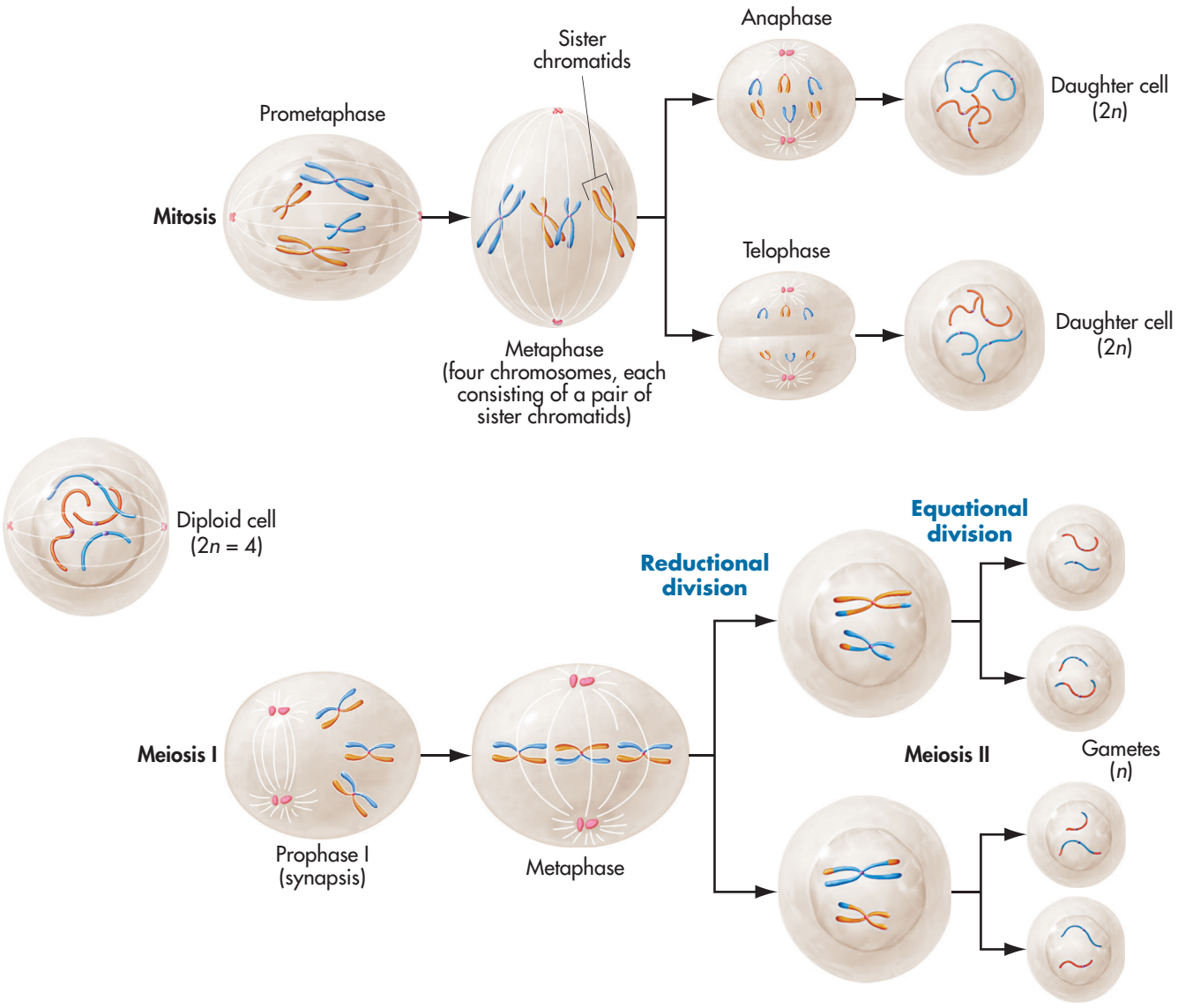


Figure 2-4 A comparison of the processes of mitosis and meiosis.

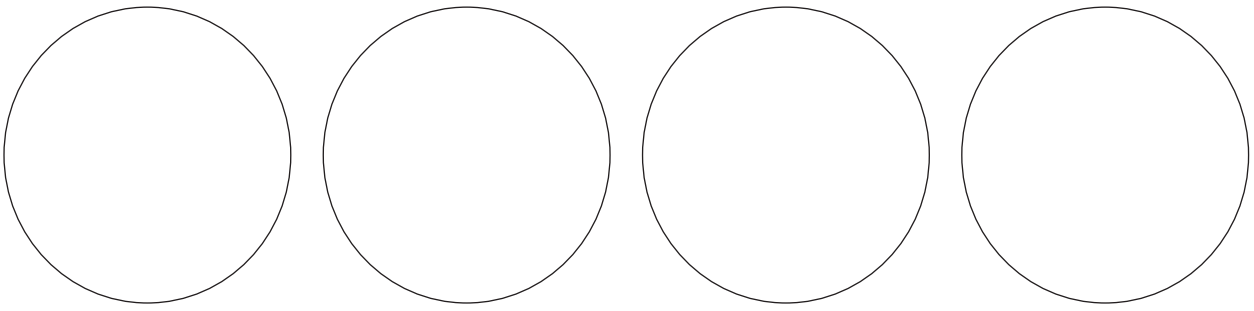
Prior to the mitotic division, the chromosomes replicate themselves, that is, they become double stranded (in a human, this process results in 46 double-stranded chromosomes). This is followed by one cell division wherein the sister chromatids separate from each other, with one strand going into each daughter cell. The end result is two daughter cells with 46 (single-stranded) chromosomes each. The full complement of chromosomes, 46, is called the **diploid** number. Thus, in mitosis, the cell begins diploid and ends diploid.

Meiosis. This process is more complicated than what occurs in mitosis. In meiosis, the genetic complement is cut in half so that the daughter cells each have half the number of chromosomes as the original cell. Because of this, meiosis is often called a *reduction division.* The genetic complement is now half the original, meaning one copy of each chromosome pair ends up in each daughter cell. This is the **haploid** number. This process produces the gametes, the sperm, and the ovum, and it takes place in the testes and ovaries, respectively. Two divisions, meiosis I and meiosis II, make this possible.

Prior to meiosis, the chromosomes replicate themselves, again becoming double stranded. Thus, there are 46 double-stranded chromosomes when meiosis I begins. During meiosis I, the homologous chromosomes pair with each other, join together, and intertwine. After the chromosome pairs have lined up across the center of the cell, the homologous chromosome pairs separate, one homologue moves toward one daughter cell, while the other homologue moves into the second daughter cell. The end result of meiosis I is two daughter cells with 23 double-stranded chromosomes each. In meiosis II, each daughter cell now divides, resulting in four daughter cells total. In the nucleus, the sister chromatids separate, with one strand going into each daughter cell. This division is similar to mitosis. The result is four daughter cells, with half the number of original chromosomes (23 each). In the male, this process is referred to as *spermatogenesis* and produces four sperm cells. In the female, this process is called *oogenesis* and produces one egg cell and three *polar bodies.* While the nuclear divisions provide all four cells with 23 chromosomes each, the egg cell receives all the cell contents from the cytokenesis, while the polar bodies disintegrate.

EXERCISE 3

Examine the phases of mitosis under the microscopes in the lab. Each slide captures a cell in the process of dividing. DO NOT touch the coarse focus control on the microscope or move the slide! It is OK to use the fine focus. Below, sketch the position of the chromosomes in the phases of mitosis that you see. Refer to Figure 2-4 for the phases. Label each sketch for the phase depicted: interphase, prophase, metaphase, anaphase, or telophase.



EXERCISE 4

Compare and contrast mitosis and meiosis in the human with the following matching questions.

_____ happens in the body cells
_____ produces 4 daughter cells
_____ begins with 46 chromosomes
_____ produces 2 daughter cells
_____ one nuclear division
_____ one chromosome replication
_____ happens in the testes and ovaries
_____ daughter cells have 23 chromosomes each
_____ two nuclear divisions
_____ daughter cells are diploid

a. mitosis
b. meiosis
c. both mitosis and meiosis

Recombination (Crossing Over)

During meiosis, an important event occurs that shuffles the genetic information around on the chromosomes. When the homologous chromosomes pair up and intertwine in meiosis I, the chromatids break and portions of chromatids bearing genes for the same trait are exchanged, or reshuffled, between homologous chromosomes. This is called **recombination** or **crossing over** (Figure 2-5). The end result may be that the original chromatids are carrying different alleles at that location. Crossing over allows a great amount of variability to be incorporated into the chromosome.

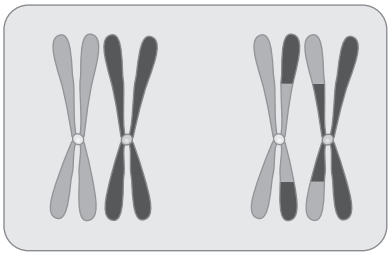


Figure 2-5 The process of recombination.

EXERCISE 5

Draw a homologous pair of chromosomes. Use one color (e.g., pink) for one member of the pair and use a second color (e.g., blue) for the second member of the pair.

Next, draw the two chromosomes crossing over, so that the two colors are touching.

Third, draw the two chromosomes after the crossing over is completed and they have shuffled their gene pairs, exchanging genes (colors) between them. Have at least one exchange.

Compare your drawing to those of others in the class, and see the amount of variation that might be possible.

DNA Structure and Function

In 1869, a chemist by the name of Friedrich Miescher found a substance in the cell nucleus that he called "nuclein." This substance became known as deoxyribonucleic acid, or DNA. In the 1950s, several researchers were attempting to discover the structure of DNA and exactly how it or some other molecule (e.g., proteins) might carry genetic information. In 1953, James Watson and Francis Crick proposed that the DNA molecule was composed of two strands that were twisted around each other in a **double helix** structure (like a twisted ladder). For their pioneering work, Watson and Crick were awarded the Nobel Prize in 1962 (Rosalind Franklin, who also worked on DNA structure, was also awarded the Nobel Prize that year; however, she died before the Nobel Prize was announced). It can be argued that the discovery of DNA as the genetic material and determination of its molecular structure are two of the most significant discoveries of the twentieth century. Understanding the function of DNA is essential for understanding life.

We now understand that DNA is made up of two chains of **nucleotides**. Each DNA nucleotide contains a phosphate, a sugar (deoxyribose sugar), and a base group. The phosphates and sugars make up the "sides" of the ladder-like structure. Each sugar is also connected to a base. The four possible bases are adenine (A), cytosine (C), guanine (G), and thymine (T). These bases join with each other in **complementary base pairs** to form the "rungs" of the ladder (Figure 2-6). The joining is very specific: A with T, C with G. Structurally and functionally, the base pairing lies at the heart of the DNA molecule. Because of this strict base pairing, the sequence of bases in the DNA molecule is preserved when the molecule is replicating or being copied.

DNA has two main functions: (1) **self-replication**, which occurs when the cell is about to divide and (2) **protein synthesis**, or the formation of proteins. Proteins are structural molecules that are important for building all the cells of the body. Proteins also act as *enzymes,* allowing necessary chemical reactions in the body to take place.

DNA replication takes place just prior to cell division and results in the chromosomes replicating themselves. Replication begins when the DNA molecule

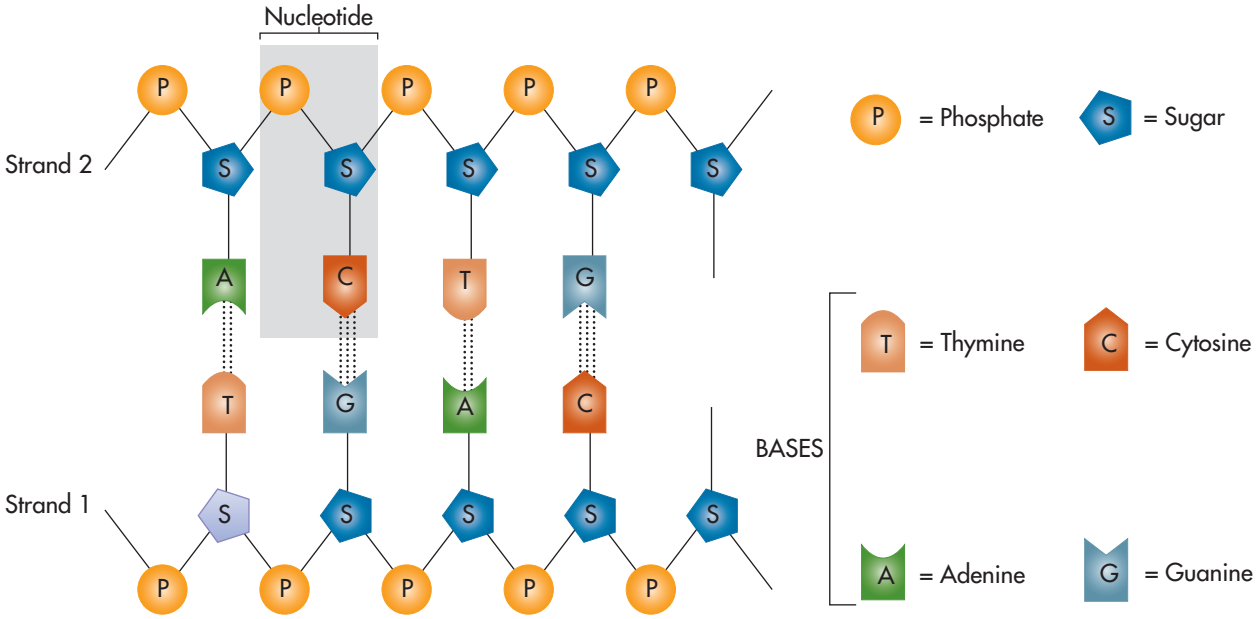


Figure 2-6 A strand of DNA demonstrating the nucleotide structure.

"unwinds" and the strands separate, then "unzips" when the hydrogen bonds between the bases are broken. This results in two single strands of DNA with exposed bases. Free DNA nucleotides within the nucleus bond to the exposed bases, according to the base pairing rules, creating two new strands of DNA. Note that each new strand contains one original strand and one new strand. DNA replication is *semiconservative*: each parental strand remains intact, while a new complementary strand is formed.

EXERCISE 6

Practice DNA base pairing:

Consider the following DNA strand: A T C C T A G G T C A G

Identify the complementary bases: _____

Now, practice DNA replication. Consider the following double-stranded DNA molecule. Notice that the DNA bases are paired accordingly. Separate the strands and replicate them, identifying which strands are original and which are the new complementary strands. Write the complementary bases for the top strand above the strand and the complementary bases for the bottom strand below the strand.

T A C G G C A A C T G A G C T

A T G C C G T T G A C T C G A

_____

_____

Protein Synthesis. The sequence of the bases in the DNA chain codes for **amino acids**, which link together to form proteins. The process of protein synthesis occurs in two stages: (1) **transcription** and (2) **translation** (Figure 2-7). First, in transcription, the DNA must be copied into a form that is able to exit the nucleus

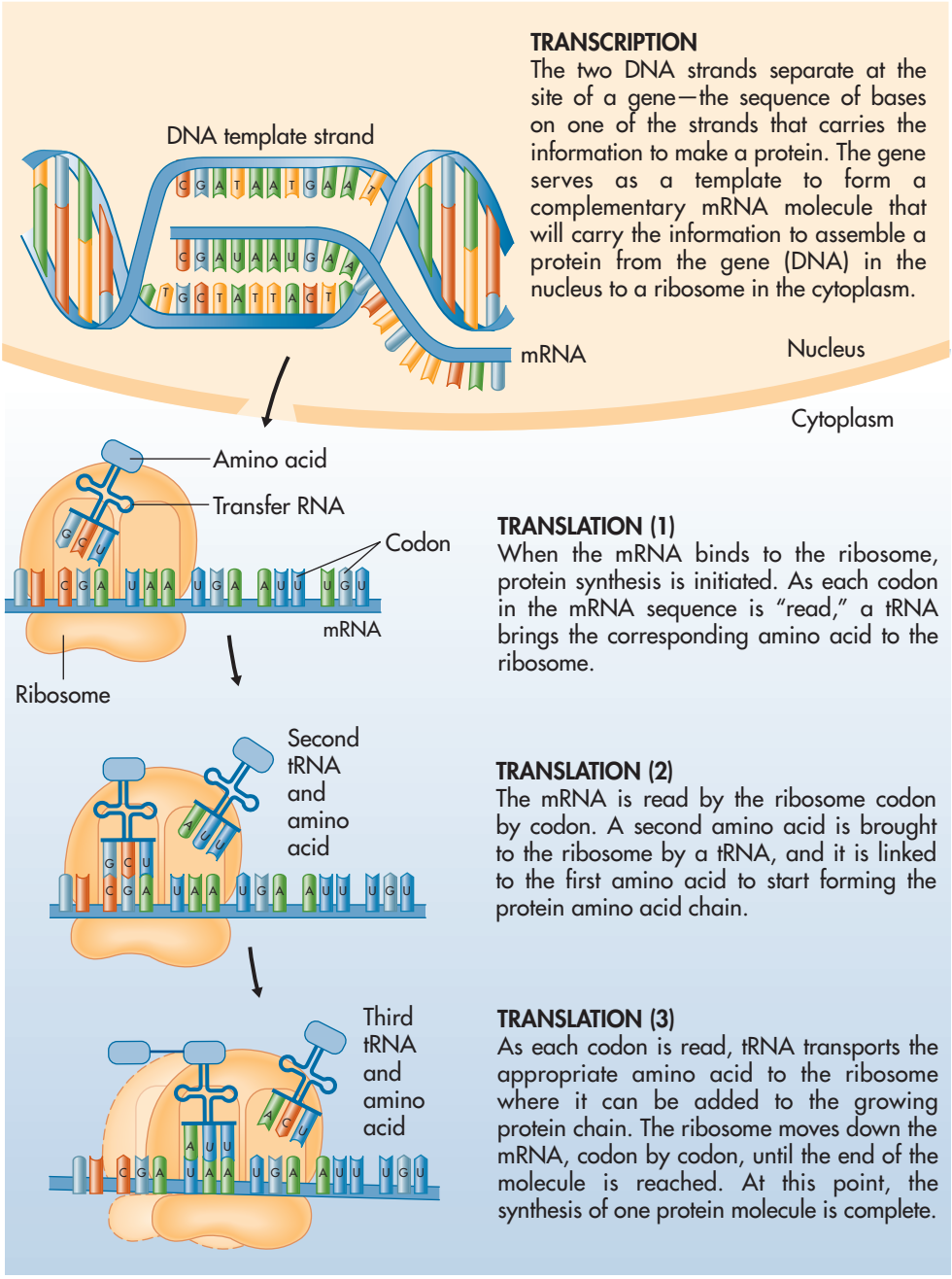


Figure 2-7 Protein Synthesis.

of the cell. This is accomplished by the formation of another type of nucleic acid, **RNA** (ribonucleic acid). RNA is a single-stranded molecule, composed of RNA nucleotides linked together. An RNA nucleotide is composed of a phosphate, a ribose sugar, and a base group. Note that the sugar molecules in DNA and RNA are slightly different. There are also differences in the bases. RNA does contain A, C, and G, but it does not contain T. Instead, RNA has a base called *uracil* (U). The base pairing is similar: C with G and A with U. The process begins with the DNA molecule unwinding and "unzipping," wherein the DNA strands again become separated. One DNA strand will be the *template strand,* the strand that is copied. This time, free RNA nucleotides in the nucleus will pair up with the exposed DNA bases on the template strand, following the base pairing rules. The strand of RNA that is formed is called *messenger RNA* or **mRNA** because it carries the DNA "message" or code. The mRNA strand may now exit the nucleus

and head out to a specific organelle in the cytoplasm of the cell, called the **ribosome**, which is the site of protein synthesis. The DNA strands pair up again.

In the second stage, *translation,* the mRNA code is scanned through the ribosome, where it is "read" like a bar code in a supermarket. "To translate" means to change from one language to another. In this process, the mRNA code is translated into a strand of amino acids that will eventually form a protein. The mRNA strand is read three bases at a time. A series of three bases in RNA is called a **codon**. The *transfer RNA,* **tRNA**, carries an amino acid and sits at the ribosome, where the tRNA anticodons will pair up with the mRNA codons. Each codon calls for one amino acid. Think of a codon like a three-letter word. As the codons are read by the tRNA, the appropriate amino acids are linked together in a long peptide chain. Many peptide chains linked together form a protein.

EXERCISE 7

The following chart lists all possible mRNA codons and the 20 amino acids they code for. These codons are scanned through the ribosome where they are "read" and matched up with complementary tRNA anticodons, which carry the amino acids. For example, the very first mRNA codon listed, UUU, codes for the amino acid phenylalanine. When the MRNA codon pairs up with the tRNA anticodon carrying phenylalanine, the first amino acid is put into place on the chain. Note that there is some redundancy in the code. Also note that some codons code for *start* or *stop*, which tells the cell where to start or stop making the protein. Using this information, fill in the blanks below the chart for the amino acid each codon calls for.

UUU Phenylalanine	UCU Serine	UAU Tyrosine	UGU Cysteine
UUC Phenylalanine	UCC Serine	UAC Tyrosine	UGC Cysteine
UUA Leucine	UCA Serine	UAA Stop	UGA Stop
UUG Leucine	UCG Serine	UAG Stop	UGG Tryptophan
CUU Leucine	CCU Proline	CAU Histidine	CGU Arginine
CUC Leucine	CCC Proline	CAC Histidine	CGC Arginine
CUA Leucine	CCA Proline	CAA Glutamine	CGA Arginine
CUG Leucine	CCG Proline	CAG Glutamine	CGG Arginine
AUU Isoleucine	ACU Threonine	AAU Asparagine	AGU Serine
AUC Isoleucine	ACC Threonine	AAC Asparagine	AGC Serine
AUA Isoleucine	ACA Threonine	AAA Lysine	AGA Arginine
AUG Start (Methionine)	ACG Threonine	AAG Lysine	AGG Arginine
GUU Valine	GCU Alanine	GAU Aspartic Acid	GGU Glycine
GUC Valine	GCC Alanine	GAC Aspartic Acid	GGC Glycine
GUA Valine	GCA Alanine	GAA Glutamic Acid	GGA Glycine
GUG Valine	GCG Alanine	GAG Glutamic Acid	GGG Glycine

Source: Laidlaw and Kopple (1987)

UCA _____ GUA _____

UGG _____ AGA _____

CUC _____ GCC _____

CAU _____ AUG _____

EXERCISE 8

The following is a template strand of DNA:

A C G G T T C A T G C A

a. What is the complementary mRNA strand?

_____

b. What are the complementary tRNA anticodons?

_____

c. Using the chart from Exercise 6, what is the sequence of amino acids for this peptide chain? *Be sure to use the mRNA codons when reading the chart!* _____

_____

_____

Section _____ ID#_____ Name: _____

Post-Lab Questions

1. Describe the difference between the autosomes and the sex chromosomes. _____

2. How many chromosomes were there in your karyotype set? Was this the normal number for humans?

3. Referring to your lecture textbook, or the Internet, discuss the clinical symptoms associated with any anomaly you identified in your karyotype. _____

4. How do you determine the sex of an individual when examining his or her karyotype?

5. How are the different types of chromosomes identified for a karyotype? _____

6. If the chromosome number for an organism is 22 before mitosis, what is the chromosome number of each daughter cell after mitosis has taken place? _____

7. Why is the DNA replicated prior to mitosis? _____

8. What do you think might happen if a cell underwent mitosis but not cytokinesis? _____

9. If a cell in an organism had 16 chromosomes before meiosis, how many chromosomes would exist in each nucleus after meiosis? What is the diploid number? What is the haploid number?

10. From a genetic standpoint, what is the significance of fertilization?

11. Describe the differences between haploid and diploid cells and where are they found.

12. Discuss the differences you observed when comparing your crossing over diagram to others in the class. How many different combinations did you see?

13. What does it mean when we say DNA replication is semiconservative?

14. Describe the differences in DNA and RNA structure.

15. To transcribe means "to make a copy of." Is an exact copy of DNA made during the process of transcription? Why or why not?

16. Where does transcription happen? What about translation? _____

_____

_____

_____

17. What amino acid would be produced if transcription took place from the DNA sequence CAT?

_____

a. If a genetic mistake took place during replication and the new DNA strand has the sequence CAG, what amino acid would result? _____

_____

b. What if the genetic mistake resulted in a DNA strand with the sequence GAT? _____

_____

c. Explain these results. _____

_____

_____

_____

Chapter 3

Principles of Inheritance

objectives

After completing this chapter you should be able to:

1. explain the difference between genotype and phenotype, homozygous and heterozygous, dominant and recessive;
2. determine the gametes produced from given genotypes and the genotypes formed from specific gamete pairs;
3. understand Mendel's Laws;
4. solve problems using Punnett squares involving monohybrid and dihybrid crosses, including the probabilities of offspring genotypes and phenotypes; and
5. understand the differences among complete dominance, incomplete dominance, codominance, and sex-linked inheritance.

Reading Questions You should read through the text of this lab prior to your class. Then answer the following questions to be ready for the classroom.

1. Mendel published his works in:
   a. 1965,
   b. 1956,
   c. 1776,
   d. 1865.

2. In a dihybrid cross:
   a. only one trait is considered,
   b. the parents are always heterozygous,
   c. two traits are considered,
   d. all offspring must be heterozygous.

3. When all self-fertilized offspring display the same traits as their parents, we know that the parents are:
   a. hybrids,
   b. heterozygous,
   c. codominant,
   d. true breeders.

4. The physical characteristics of an organism are referred to as the:
   a. dominant allele,
   b. gametes,
   c. phenotype,
   d. genotype.

5. An individual who is carrying two of the same alleles for a gene is known as:
   a. homozygous,
   b. heterozygous,
   c. dominant,
   d. a hybrid.

6. The allele that is masked or hidden in the genotype is the:
   a. heterozygote,
   b. recessive allele,
   c. dominant allele,
   d. true breeder.

7. When both alleles are fully expressed in the phenotype, this is called:
   a. incomplete dominance,
   b. codominance,
   c. sex-linked,
   d. recessive.

8. **True or False:** Sex-linked traits are often located on the X chromosome.

9. **True or False:** Mendel's Law of Independent Assortment states that during meiosis, the chromosome pairs separate, so that each newly formed gamete receives one chromosome from each pair.

10. **True or False:** Numerous traits in humans are inherited in a simple Mendelian fashion.

The Basic Principles of Inheritance

Gregor Mendel was an Austrian monk who discovered the basic principles of inheritance that we use today. He studied mathematics, botany, and physics at the University of Vienna before beginning his experiments, breeding the common garden pea, in 1856. He published his work in 1865, but unfortunately the work was unrecognized and unappreciated at the time. Mendel was not the first person to study heredity in plants, but he was the first person to use statistics in his analyses. Working in the monastery garden, Mendel followed the inheritance of simple traits through several generations. He discovered that the information that controlled inheritance behaved like particles that were passed from one generation to the next. Today, we call these particles *genes*.

Mendel concluded that each pea plant contained two particles of information for each trait. Each parent plant contributed one particle of information for each trait to each of its offspring.

Can you explain this discovery using our modern understanding of cell biology and DNA from Chapter 2? _____

_____

_____

_____

The common garden pea has both male and female parts that can breed with each other in the same flower and also are able to self-fertilize. To begin his experiments, Mendel chose plants that were *true-breeders,* meaning that all self-fertilized offspring displayed the same traits as their parents. For example,

if a true-breeding plant with round seeds self-fertilized, then all offspring also would have round seeds. In other words, the plant carries the same allele for the gene at each locus (i.e., each plant carries the round allele for the seed shape gene). When parents that are true-breeders for different traits are mated (e.g., round seeds and wrinkled seeds), the offspring are called **hybrids**. The offspring have alleles for both traits (e.g., round seeds and wrinkled seeds). The **genotype** is the genetic constitution of an organism, the actual alleles present. The **phenotype** describes the physical characteristics of an organism, the expression of its genes. Mendel was breeding plants, examining their phenotypes, and trying to infer the underlying genotypes.

When the transmission of one trait at a time is studied, it is called a **monohybrid cross**. When two traits at a time are studied, it is called a **dihybrid cross**. Beginning with the monohybrid cross, recall that somatic cells are diploid: They contain two copies of each chromosome arranged in pairs. These homologous pairs carry the same genes, although they may have different alleles. For one trait that has two forms (e.g., seed shape in pea plants has two forms: round seeds and wrinkled seeds), three combinations of alleles are possible. Different types of notation are used to represent the alleles; a common notation is used here. Using *R* to indicate the allele for round seeds, and *r* to indicate the allele for wrinkled seeds, if both chromosomes carry the *R* allele the plant is *RR,* while if both chromosomes carry the *r* allele the plant is *rr.* However, if one chromosome carries the *R,* while the other member of the pair carries the *r,* then the gene pair is *Rr. Remember*: Sex cells are haploid and only carry *one* of these alleles. So the sperm and ovum would carry either an *R* or an *r*.

EXERCISE 1

a. Mendel's pea plants carry two alleles for the flower color gene: *P* for purple flowers and *p* for white flowers. What three possible combinations might exist in any one plant?

_____

b. Mendel's pea plants also carry two genes for plant height: *T* for tall plants and *t* for short plants. Consider the genotypes in the following table and indicate the possible genotypes of the gametes (*Remember*: Each gamete will only carry one allele).

Diploid Genotype	Gamete Genotype	Gamete Genotype
TT		
Tt		
tt		

c. Working in the opposite direction, during fertilization when the sperm and egg fuse, the haploid cells come together and form a diploid zygote. In the table given below, provide the diploid genotypes that would occur by fusion of the following gamete genotypes.

Gamete Genotype (Sperm)	Gamete Genotype (Ovum)	Diploid Genotype (Zygote)
t	*t*	
T	*t*	
T	*T*	

In the preceding examples, there are two alleles for each gene. A plant whose genotype is *RR* or *Rr* has round seeds. *RR* and *Rr* are the genotypes, and "round" is the phenotype. This is the **dominant** condition. Dominance is the ability of one allele to mask or hide the expression of the other allele. In this case round (*R*) is dominant to wrinkled (*r*). The **recessive** allele is the allele that is not expressed, the one that is hidden, in this case *r*, wrinkled. A plant with two copies of the recessive allele, *rr*, would exhibit wrinkled seeds. The dominant gene is always written as a capital letter, while the recessive gene is written as a lowercase letter. These are cases of *complete dominance.* When both alleles are identical, the individual is said to be **homozygous** for that trait (*RR* or *rr*). When both copies are the dominant gene (*RR*), the individual is *homozygous dominant.* When both alleles are for the recessive condition (*rr*), the individual is said to be *homozygous recessive.* When the alleles are different (*Rr*), then the individual is said to be **heterozygous**.

EXERCISE 2

We know that purple flowers in pea plants are dominant to white flowers. Using the example for flower color in Exercise 1, identify the flower colors for plants that have the following genotypes. Label the homozygous and heterozygous conditions.

- *PP* _____
- *Pp* _____
- *pp* _____

Why? _____

_____

EXERCISE 3

In Mendel's pea plants, yellow seeds are dominant to green seeds. Using *Y* for yellow and *y* for green, list the three possible genotypes, followed by their phenotype (yellow or green), and label the homozygous and heterozygous conditions.

_____

_____

_____

Mendel's Laws of Inheritance

From the basic genetic principles already described, Mendel formulated two laws (or principles), which are described here using modern terminology:

- **Law of Segregation:** During meiosis, the chromosome pairs separate, so that each newly formed gamete receives one chromosome from each pair (i.e., one allele from each pair).
- **Law of Independent Assortment:** During meiosis, the members of different pairs of alleles assort independently into gametes (especially so if they are on different chromosomes). In other words, the segregation of one

pair of chromosomes does not influence the segregation of another pair of chromosomes in the same sex cells.

For example, we know that a pea plant with the genotype *Rr* produces the gametes *R* and *r*. If we consider an example where two traits are being compared at one time, and this same plant was also *Tt,* then all possible combinations of gametes would be *RT, Rt, rT,* and *rt.*

EXERCISE 4

For this exercise, let's expand our practice to include three traits at once.

a. What gametes are produced from a plant that is *Pp*? _____

_____

b. What gametes are produced from a plant that is *PpTt*? _____

_____

c. What gametes are produced from a plant that is *PpTtYy*? _____

_____

Punnett Squares

The **Punnett square** is used to figure out all possible results of mating between individuals. In this way, we examine the gametes in all possible combinations. We are then able to determine the probabilities of offspring genotypes and phenotypes. To practice a Punnett square, start with the parental gametes along the top and left side of the square and then cross-multiply.

For example, the gene for seed color has two alleles, *Y* and *y*. Let *Y* stand for the yellow allele, and *y* stand for the green allele, where yellow is dominant to green. If one parent is homozygous dominant (*YY*) and the other parent is a heterozygote (*Yy*), a Punnett square is calculated with the alleles for one parent along the top, and the alleles for the other parent along the left side. The possible offspring may be calculated by cross-multiplying as follows:

	Y	*Y*
Y	*YY*	*YY*
y	*Yy*	*Yy*

The possible genotypes formed from the mating are *YY* and *Yy*. Of the offspring, 50% (2 out of 4) are homozygous *YY,* and 50% of the offspring are heterozygous *Yy* (2 out of 4). There are no homozygous recessives (*yy*) produced by this mating. All offspring produced from this mating are yellow due to dominance. This is a simple monohybrid cross.

Let's look at a second example using plant height. This gene also has two alleles: *T* for tall and *t* for short. If one parent is a short plant (homozygous recessive) and the other parent is homozygous for tall, the offspring would be:

	t	*t*
T	*Tt*	*Tt*
T	*Tt*	*Tt*

In this case, all the offspring (100%) are heterozygous for the *Tt* genotype. Due to dominance, all the offspring are tall.

EXERCISE 5

a. Considering that purple is dominant to white, and using *P* for purple and *p* for white, draw a Punnett square crossing a heterozygous purple-flowered plant and a white-flowered plant.

b. What are the genotypes produced by this mating? _____

c. What are the phenotypes produced by this mating? _____

d. What percentage of the offspring is homozygous? _____

EXERCISE 6

a. We know that the allele for round seeds is dominant to the allele for wrinkled seeds. Using *R* for round and *r* for wrinkled, draw a Punnett square crossing two plants that are both heterozygous.

b. What percentage of the offspring is homozygous dominant? _____

c. What percentage of the offspring is homozygous recessive? _____

d. What percentage of the offspring is heterozygous? _____

e. What percentage of the offspring is wrinkled? _____

f. What percentage of the offspring is round? Why? _____

Figure 3-1 A roan cow.

Incomplete Dominance, Codominance, and Sex-Linked Inheritance

The preceding examples illustrate complete dominance. Cases of incomplete dominance, codominance, and sex-linked inheritance also occur. **Incomplete dominance** describes a situation wherein one allele is not completely dominant over another. It generally allows a form of blending to occur. An excellent example is seen in the flower color of petunias. Red-flowered petunias are *RR,* white-flowered petunias are *WW*, and the heterozygous *RW* has pink flowers. Notice the different notation used here. Because neither allele is completely dominant to the other, we do not use a lowercase letter and the resulting offspring may have a different phenotype from the parents (pink). The following Punnett Square calculates the offspring from a mating of a red- flowered plant to a white-flowered plant:

	R	*R*
W	*RW*	*RW*
W	*RW*	*RW*

Note that all offspring are *RW*, pink-flowered plants.

In **codominance**, both alleles are fully expressed in the phenotype. One example is the coat color of shorthorn cattle. Red shorthorns are *RR,* while white shorthorns are *WW.* In this case the heterozygote *RW* has a reddish-gray coat and is called a roan. The roan color is due not to incomplete dominance or blending but to the animal having *both* red and white hairs (Figure 3-1). A Punnett Square crossing a red-coated animal with a white-coated animal would look identical to the Punnett Square above for petunias, but would be interpreted differently in terms of color.

EXERCISE 7

a. Calculate the Punnett square crossing two pink-flowered petunias. What percentage of the offspring are pink, white, and red?

b. Calculate the Punnett square crossing a roan shorthorn bull with a roan cow. What percentage of the offspring has a roan coat, a red coat, and a white coat?

Recall that in many organisms, including humans, sex is determined by the sex chromosomes. Human females are *XX,* while human males are *XY* at their 23rd chromosome pair. The genes that occur on the sex chromosomes are considered **sex-linked**. The X chromosome is large and contains many genes, thus most sex-linked traits are located on the X chromosome. In humans there are two genes—color vision and hemophilia (a blood clotting disease)—that are found on the X chromosome and not on the Y chromosome, making them sex-linked.

Normal color vision is dominant over red-green color blindness (a situation in which red and green cannot be differentiated). Due to complete dominance, in order for a female to be affected by color blindness, both her X chromosomes must carry the color-blind allele. A female carrying only one copy of the color-blind allele has normal vision and is called a **carrier**. However, a male only has one X chromosome. If his single X carries the allele for color blindness, he will be color-blind.

EXERCISE 8

a. Using X for normal vision and X^C for color-blind, calculate the Punnett square that would result from a carrier female mating with a color-blind male. *Hint:* The female genotype is X^CX and the male genotype is X^CY.

b. What are the chances that they will produce a color-blind son? _____

c. What are the chances that they will produce a color-blind daughter? _____

d. Is there any way these parents might produce a daughter with normal vision? If so, how? _____

e. Is there any chance these parents might produce a son with normal vision? If so, how? _____

Mendelian Traits in Humans

In the preceding pages, we have observed many examples of traits in the common pea plant that are inherited through a simple dominant/recessive relationship. Most human traits are very complex and are affected by several genes (*polygenic*). Examples of polygenic inheritance are hair color, eye color, intelligence, and musical ability. However, several traits have been identified in humans that represent simple inheritance as defined by Mendel. The *Online Mendelian Inheritance of Man* Web site is very helpful for identifying and describing those traits that are inherited in a simple fashion in humans (www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM). The following is a list of ten common traits in humans that are inherited in a Mendelian (simple) fashion:

1. **Mid-digital hair.** Examine the middle segment of your fingers for the presence of hair, which is inherited as a dominant (*MM, Mm*). A complete absence of hair represents the homozygous recessive condition (*mm*).
2. **Tongue rolling.** The ability to roll the sides of the tongue up into a tube shape, inherited as a dominant (*RR, Rr*). Inability to roll the tongue is due to the recessive condition (*rr*).
3. **Earlobe attachment.** Most people have free earlobes, which hang down from the head and are inherited as a dominant (*FF, Ff*). Homozygous recessive individuals (*ff*) have earlobes that are attached directly to the head.
4. **Hitchhiker's thumb.** If you can bend your thumb back at an angle of greater than 45 degrees, you have the recessive condition (*hh*). Individuals who are *HH* or *Hh* do not have Hitchhiker's thumb. Because this trait is a recessive, two copies are needed to display the phenotype.
5. **Ability to taste PTC.** Phenyl-thio-carbimide is a bitter, synthetic chemical similar to a substance found in kale, turnips, and brussel sprouts. The ability to taste PTC is inherited as a dominant. About 70% of Americans can taste PTC.
6. **Darwin's tubercle.** This is a thickening, or projection, on the rim of the ear due to a thickening of cartilage. It is inherited as a dominant.
7. **Interlocking fingers and thumbs.** Placing the left thumb over the right is the dominant condition. Switching your thumbs feels awkward and uncomfortable.
8. **Earwax.** Flaky, grayish-white earwax is the recessive condition, while sticky, yellowish earwax is the dominant condition.
9. **Palmaris longus tendon.** The presence of two tendons on the inside of your wrist is inherited as a recessive. The dominant condition exhibits a third centrally located tendon (palmaris longus).
10. **Cleft (dimpled) chin.** The presence of a dimple in the chin is inherited as a dominant trait.

You can also calculate Punnett Squares for these traits. For example, if a man who is homozygous dominant for Hitchhiker's thumb (meaning he does not have the ability) marries a woman who is heterozygous for the condition (and also does not have the ability), the possible combinations for their offspring are:

	H	*H*
H	*HH*	*HH*
h	*Hh*	*Hh*

In this case, all the offspring carry the dominant allele, meaning they cannot bend their thumbs backward to a great degree. Neither the parents, nor the offspring, will exhibit Hitchhiker's thumb.

EXERCISE 9

a. A trait which is inherited as a dominant in humans is the ability to taste a bitter substance called PTC. Using *P* for PTC tasting and *p* for nontasters, draw a Punnett square crossing a heterozygous taster and a nontaster.

b. What percentage of the offspring is homozygous dominant? _____

c. What percentage of the offspring is homozygous recessive?

d. What percentage of the offspring are tasters?

EXERCISE 10

a. The consistency of a person's earwax is also inherited in a simple dominant/recessive fashion. Using *E* to indicate the dominant sticky, yellowish phenotype and *e* to indicate the recessive gray, flaky phenotype, draw a Punnett square crossing a homozygous dominant person and a heterozygous person.

b. What are the chances of getting a child with the flaky gray earwax? _____

c. What percentage of the offspring is heterozygous? _____

d. What percentage of the offspring is homozygous? _____

EXERCISE 11

For each trait in the following chart, list your phenotype. Using the information collected by everyone in your class, fill in the rest of the chart.

Trait	Your Phenotype Dom or Rec	# Dominant in Class	# Recessive in Class	% Dominant	% Recessive
Mid-digital hair					
Tongue rolling					
Earlobes					
Hitchhiker's thumb					
PTC tasting					
Darwin's tubercle					
Interlocking fingers/thumbs					
Earwax					
Palmaris longus					
Cleft (dimpled) chin					

Dihybrid Crosses

All the examples and problems presented so far have demonstrated crossing one trait at a time. In this section, we will work with crossing two traits at once: a *dihybrid cross.* To make things easier, we are assuming that the traits are *not* carried on homologous chromosomes. Let's work with two of the traits previously listed: tongue rolling and cleft (dimpled) chin. Let *T* stand for the dominant condition and *t* for the recessive condition in tongue rolling, and let *D* stand for the dominant condition (dimpled) and *d* for the recessive condition (no dimple) for cleft chin.

EXERCISE 12

a. List all possible genotypes for an individual with the ability to roll the tongue and a cleft chin. (*Hint:* Four combinations are possible.)

_____

_____

b. List all possible combinations for an individual who cannot roll his or her tongue and has a cleft chin. _____

_____

_____

c. List all possible genotypes for an individual who can roll his or her tongue and does not have a cleft chin. _____

_____

_____

d. What is the genotype for an individual who cannot roll his or her tongue and does not have a cleft chin? _____

e. Suppose an individual is heterozygous for both traits (tongue rolling and cleft chin). What is their genotype? _____

f. What are the genotypes of the gametes this person could produce? _____

EXERCISE 13

Using the answer you determined for Exercise 12, question f, set up a Punnett square for a dihybrid problem, crossing two individuals that are heterozygous for tongue rolling and cleft chin. This square will have 16 boxes. The parental genotypes on the top and left side of the following box have been inserted to help get you started. Calculate the possible offspring genotypes. When you are finished, answer the questions below.

	TD	*Td*	*tD*	*td*
TD				
Td				
tD				
td				

a. What are the chances of having a child that is:

- *TTDD* _____
- *TtDd* _____
- *TtDD* _____
- *ttDd* _____

b. What are the chances of having a child who has a cleft chin and cannot roll his or her tongue? _____

c. What are the chances of having a child who does not have a cleft chin and can roll his or her tongue? _____

d. What are the chances of having a child who cannot roll his or her tongue and does not have a cleft chin? _____

Ethics and The Future

Mendel's pioneering experiments provide the foundation for today's genetic research and technology. Many diseases have a genetic basis—for example, cystic fibrosis, Huntington's disease, sickle-cell anemia, and familial hypercholesterolemia, among many others. Modern genetic research is treating diseases that were once thought to be incurable. Insulin and human growth hormone are now produced through genetic engineering techniques. The latest in cancer research hopes to remove cells from the bodies of cancer patients, genetically alter them to enhance their tumor resistance, and then reinsert them into the body of the patient. Genetic engineering is also common in agricultural products—for example, many crops are genetically engineered for resistance to disease, drought, and/or pests. Considerable controversy surrounds these modern techniques. Some groups strongly oppose bioengineering and any "tinkering" with the genome. To be a responsible member of our society, it is important that you are able to make an educated decision about these issues. That understanding begins here, with the material covered in this chapter.

EXERCISE 14

Cystic fibrosis is the most common metabolic error in European-derived ("white") populations, with about 1 of every 1,600 being a carrier. Individuals born with cystic fibrosis are lacking an enzyme that allows them to break down thick mucus in the lungs and makes them susceptible to serious and often fatal lung infections. With aggressive treatment, most individuals may reach adulthood; otherwise death from pneumonia is likely in childhood. There is no cure; however, genetic engineering offers promise for these affected individuals. Cystic fibrosis is inherited as a recessive, and individuals with one copy of the gene are carriers of the disease. The gene has been located on chromosome 7.

a. What percentage of the gametes of a heterozygote individual will contain the recessive allele? _____

b. Draw a Punnett square crossing two individuals who are heterozygous for this trait.

c. What percentage of the offspring will be affected with cystic fibrosis?

d. What percentage of the offspring will be normal? _____

e. What percentage of the offspring will also carry the trait? _____

f. Is it possible for an affected child to be born to one healthy parent and one carrier? _____

Section _____ ID#_____ Name: _____

Post-Lab Questions

1. Distinguish between incomplete dominance and codominance. _____

2. What does it mean when we say that a trait is sex-linked?_____

3. How might the Law of Independent Assortment be violated if two traits were on the same chromosome?

4. Mendel's pea plants carry two alleles of the gene for seed shape: *R* for round seeds and *r* for wrinkled seeds. Consider the genotypes in the following table, and indicate the possible genotypes of the gametes.

Diploid Genotype	Gamete Genotype	Gamete Genotype
RR		
Rr		
rr		

5. In the chart given below, provide the diploid genotypes that would occur by fusion of the following genotypes.

Gamete Genotype (Sperm)	Gamete Genotype (Ovum)	Diploid Genotype (Zygote)
r	*r*	
R	*r*	
R	*R*	

6. For the following, state whether the genotype is homozygous or heterozygous and whether the plant would have round or wrinkled seeds.

   a. *RR* _____

   b. *Rr* _____

   c. *rr* _____

7. Considering that the allele for tall plants is dominant to the allele for short plants, and using *T* for tall and *t* for short, draw a Punnett square crossing a heterozygous tall plant and a short plant.

a. What percentage of the offspring is homozygous dominant? _____

b. What percentage of the offspring is heterozygous? _____

c. What percentage of the offspring is homozygous recessive? _____

d. What percentage of the offspring is tall? Why?

e. What percentage of the offspring is short? Why?

8. Considering that the allele in humans for tongue rolling is dominant to the allele for non-rolling, and using *R* for rolling and *r* for non-rolling, draw a Punnett square crossing a heterozygous roller and a homozygous roller.

a. What percentage of the offspring is homozygous dominant? _____

b. What percentage of the offspring is heterozygous? _____

c. What percentage of the offspring is homozygous recessive? _____

d. What percentage of the offspring can roll their tongue? Why?

e. What percentage of the offspring cannot roll their tongue? Why?

9. Considering that the allele for a dimpled chin is dominant to the allele for a non-dimpled chin, and using *D* for dimpled and *d* for non-dimpled, draw a Punnett square crossing a homozygous person with a dimpled chin and a person without a dimpled chin.

a. What percentage of the offspring is homozygous dominant? _____

b. What percentage of the offspring is heterozygous? _____

c. What percentage of the offspring is homozygous recessive? _____