CHAPTER 15

The Human Genome

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One thing to notice about this group of hikers is that none of them look alike. The diversity of traits among the human race stems from one microscopic molecule—DNA.

HS-LS3-1, HS-LS3-2, HS-ETS1-1, HS-ETS1-2

CASE STUDY

DNA—to test or not to test?

In 1990, scientists around the world launched the Human Genome Project. The goal was to determine the complete nucleotide sequence of human DNA, and to map the location of every gene. Thirteen years later, on time and under budget, the project was completed. The scientists identified about 20,000 genes, an unexpectedly small number given the complexity of the human species.

Today, new technology allows DNA to be sequenced a lot more rapidly, and for a lot less money. Machines can now automatically sequence the human genome in a day or two for less than one thousand dollars! For much less than that, many companies will now analyze samples of your DNA to search for specific sequences that reveal ethnic ancestry and tendencies to develop certain diseases.

What has been learned from exploring the details of the human genome? One example is the way human relationships can now be traced with great precision. The genomes for all humans are similar, but individual differences show how closely two people are related, and can even solve historical mysteries. For example, the remains of Richard III, an English king who died in 1485, were identified by comparing a DNA sample with a sample from one of his living descendants. Similar studies have confirmed the fates of members of the royal family of Russia after the Russian revolution. In criminal justice, the routine use of DNA in criminal investigations has helped to identify the guilty and exonerate the innocent in many cases.

DNA sequencing also helps identify genetic diseases. An important example is Tay-Sachs disease, which destroys nerve cells in infants. About 1 in 250 people carry the allele that causes the disease, and the rate is higher among certain populations. A simple blood test is now available to couples that can determine whether either of them has the recessive disease-causing allele. As you know from basic genetics, if both parents have the allele, the probability that a child will inherit the disease is 1 out of 4. In Israel, where the disease is more prevalent than elsewhere, testing and counseling have reduced Tay-Sachs cases by nearly 90 percent.

DNA tests are now available for hundreds of genetic disorders, raising a number of important questions. Should potential parents be required to be tested for genetic disorders? If such tests show a probability that two potential parents might produce children with a genetic disorder, does a couple have the right to take a chance and have children anyway? Does government have a role in trying to eliminate these tragic and costly disorders from the population?

Throughout this chapter, look for connections to the CASE STUDY to help you answer these questions.

LESSON 15.1

Human Chromosomes

KEY QUESTIONS

- *How are human karyotypes used?*
- *What patterns of inheritance do human traits follow?*
- *How can pedigrees be used to analyze human inheritance?*

HS-LS3-1: Ask questions to clarify relationships about the role of DNA and chromosomes in coding the instructions for characteristic traits passed from parents to offspring.

VOCABULARY

genome
karyotype
sex chromosome
autosome
sex-linked gene
pedigree

READING TOOL

Before you read the lesson, reflect on what you know and what you want to know about human genetics and chromosomes. Fill in the first two columns of the chart in your **Biology Foundations Workbook.** After you read, fill in the third column about the things you have learned during the reading.

If you had to pick an ideal animal for the study of genetics, you certainly wouldn't pick one that produced very few offspring, had a long life span, and could not be grown in a lab. Yet, when we study human genetics, this is exactly the sort of organism we deal with. Given all of these difficulties, it may seem a wonder that we know as much about human genetics as we do.

Karyotypes

What makes us human? We might try to answer that question by looking under the microscope to see what is inside a human cell. Not surprisingly, human cells look much like the cells of other animals. To find what makes us uniquely human, we have to look deep into the genetic instructions that build each new individual. This means that we have to explore the human genome. A **genome** is the full set of genetic information that an organism carries in its DNA.

The analysis of any genome starts with chromosomes, which are bundles of DNA and protein found in the nuclei of eukaryotic cells. To see human chromosomes clearly, cell biologists photograph cells in mitosis, when the chromosomes are fully condensed and easy to view. Scientists then arrange images of each chromosome to produce a **karyotype** (KAIR ee uh typ). ***A karyotype shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size.***

The karyotype shown in **Figure 15-1** is from a typical human cell, which contains 46 chromosomes, arranged in 23 pairs. Why do our chromosomes come in pairs? Remember that the first cell of a human is formed when a haploid sperm, carrying 23 chromosomes, fertilizes a haploid egg, also with 23 chromosomes. The resulting diploid cell develops into a new individual and carries the full complement of 46 chromosomes—two sets of 23.

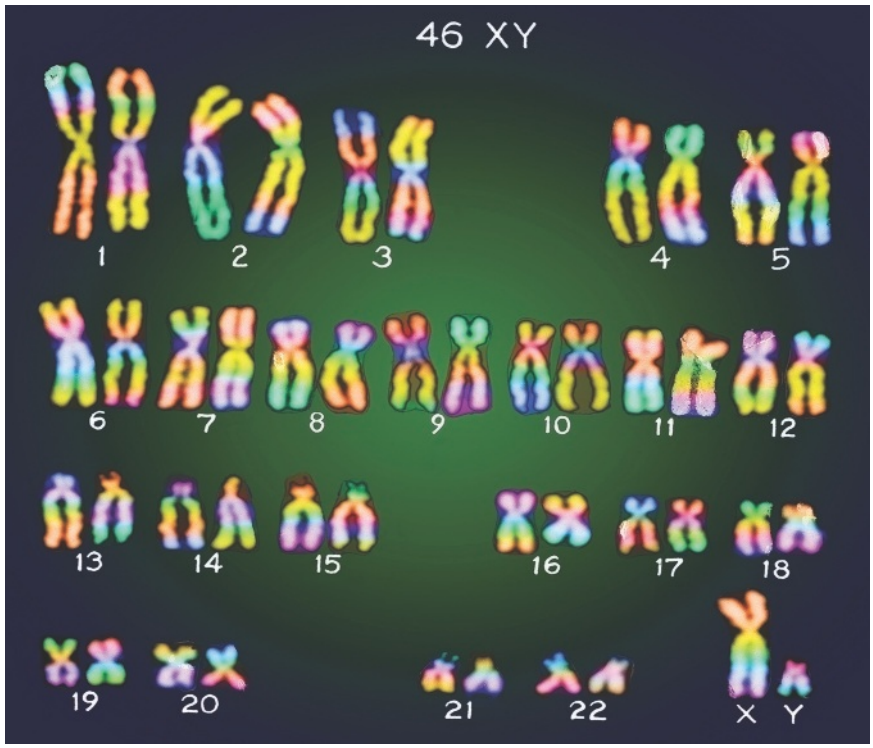


Figure 15-1

Human Karyotype

A typical human cell has 23 pairs of chromosomes. These color-enhanced images of a complete set of chromosomes have been arranged to form a karyotype.

Sex Chromosomes

Two of the 46 chromosomes in the human genome are known as **sex chromosomes**, because they determine an individual's sex. The two sex chromosomes in humans are shown in **Figure 15-2**. Females have two copies of the X chromosome. Males have one X chromosome and one Y chromosome.

All human egg cells carry a single X chromosome (23,X). However, half of all sperm cells carry an X chromosome (23,X) and half carry a Y chromosome (23,Y). This means that there is a 50 percent probability that an egg cell will be fertilized by an X-carrying sperm, and a 50 percent probability that it will be fertilized by a Y-carrying sperm. As a result, just about half the zygotes will be males and half will be females.

More than 1400 genes are found on the X chromosome. The Y chromosome, which is smaller, contains only about 158 genes, many of which are associated with male sex determination and sperm development.

Autosomal Chromosomes

To distinguish them from the sex chromosomes, the remaining 44 human chromosomes are known as autosomal chromosomes, or **autosomes**. The complete human genome consists of 46 chromosomes, including 44 autosomes and 2 sex chromosomes. To quickly summarize the total number of chromosomes present in a human cell—both autosomes and sex chromosomes—biologists write 46,XX for females and 46,XY for males.

READING CHECK Apply Concepts Why are males and females born in a roughly 50:50 ratio?

Figure 15-2

X and Y Chromosomes

The human X chromosome contains roughly ten times as many genes as the human Y chromosome. The figure shows the locations of some genes on each of the sex chromosomes.

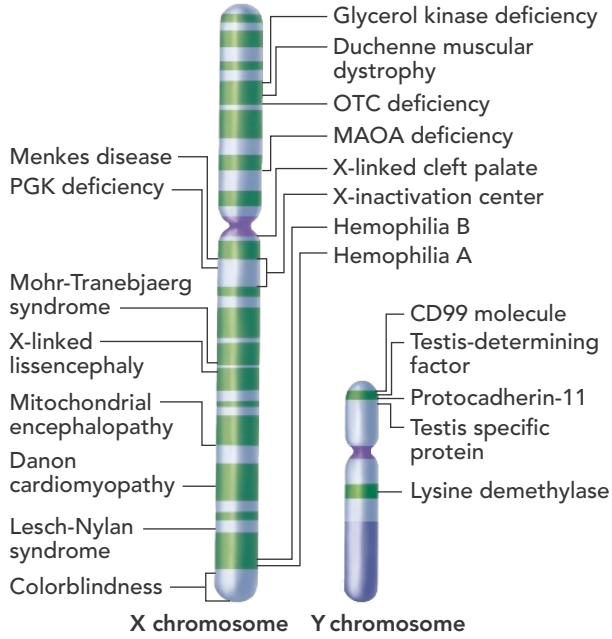


Figure 15-3

Recessive Alleles

Some recessive alleles in humans produce variations, like red hair or type O blood. Other alleles result in harmful traits, like Tay-Sachs disease and cystic fibrosis.

INTERACTIVITY

Determine which set of parents may have children with colorblindness.

Transmission of Human Traits

The study of human genetics has progressed rapidly in recent years because of the use of molecular techniques for studying DNA. What have these studies shown? Human genes follow the same patterns of inheritance as the genes of other organisms.

Dominant and Recessive Alleles

Many human traits follow a pattern of simple dominance. For instance, a gene on chromosome 16 known as *MC1R* helps determine skin and hair color. Some of *MC1R*'s recessive alleles produce red hair, as in **Figure 15-3**. An individual with red hair usually has two of these recessive alleles, inheriting a copy from each parent. Dominant alleles for the *MC1R* gene help produce darker hair colors.

Another trait that displays simple dominance is the Rhesus, or Rh blood group. The allele for Rh factor comes in two forms: Rh^+ and Rh^-. Rh^+ is dominant, so an individual with both alleles (Rh^+/Rh^-) is said to have Rh positive blood. Rh negative blood is found in individuals with two recessive alleles (Rh^-/Rh^-).

Codominant and Multiple Alleles

The alleles for many human genes display codominant inheritance. One example is the ABO blood group, determined by a gene on chromosome 9 with three alleles: I^A, I^B, and i. Alleles I^A and I^B are codominant. They produce molecules known as antigens on the surface of red blood cells. As **Figure 15-4** shows, individuals with alleles I^A and I^B produce both A and B antigens, making them blood type AB. The i allele is recessive. Individuals with alleles I^AI^A or I^Ai produce only the A antigen, making them blood type A. Those with I^BI^B or I^Bi alleles are type B. Those homozygous for the i allele (ii) produce no antigen and are said to have blood type O. If a patient has AB-negative blood, it means the individual has I^A and I^B alleles from the ABO gene and two Rh^- alleles from the Rh gene.

Figure 15-4

Human Blood Groups

Analyze the data in the table to see the relationship between genotype and phenotype for the ABO blood group. The table also shows which blood types can safely be transfused into people with other blood types.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	I^AI^A or I^Ai	A	A, AB	A, O
B	I^BI^B or I^Bi	B	B, AB	B, O
AB	I^AI^B	A and B	AB	A, B, AB, O
O	ii	None	A, B, AB, O	O

Sex-Linked Inheritance

Because the X and Y chromosomes determine sex, the genes located on them show a pattern of inheritance called sex-linkage. A **sex-linked gene** is a gene located on a sex chromosome. As you might expect, genes on the Y chromosome are found only in males and are passed directly from father to son. Genes located on the X chromosome are found in both sexes, but the fact that men have just one X chromosome leads to some interesting consequences.

For example, humans have three genes responsible for color vision, all located on the X chromosome. In males, a defective allele for any of these genes results in colorblindness, an inability to distinguish certain colors, like those shown in **Figure 15-5**. The most common form, red-green colorblindness, occurs in about 1 in 12 males. Among females, however, colorblindness affects only about 1 in 200. Why is there such a difference? In order for a recessive allele, like colorblindness, to be expressed in females, it must be present in two copies—one on each of the X chromosomes. This means that the recessive phenotype of a sex-linked genetic disorder tends to be much more common among males than among females.

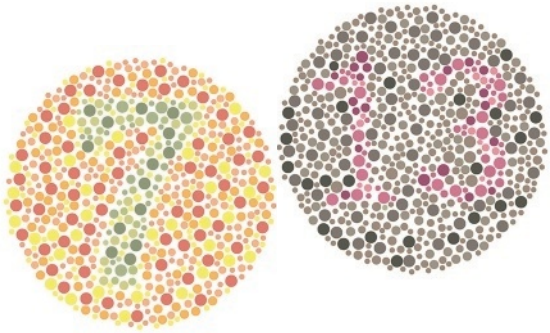


Figure 15-5

Eye Test

Doctors use images like these to test for colorblindness. A person who is colorblind will perceive only random dots, and not the colored numbers inside the circles.

X-Chromosome Inactivation

If just one X chromosome is enough for cells in males, how does the cell "adjust" to the extra X chromosome in female cells? The answer was discovered by British geneticist Mary Lyon. In female cells, most of the genes in one of the X chromosomes are inactivated, forming a condensed region in the nucleus known as a Barr body. A special RNA molecule binds to the inactivated chromosome and keeps it in the condensed state.

The same process happens in other mammals. In cats, for example, a gene that controls the color of coat spots is located on the X chromosome. One X chromosome may have an allele for orange spots and the other X chromosome may have an allele for black spots. In cells in some parts of the body, one X chromosome is switched off. In other parts of the body, the other X chromosome is switched off. As a result, the cat's fur has a mixture of orange and black spots, like those in **Figure 15-6**. Male cats, which have just one X chromosome, can have spots of only one color. Therefore, if the cat's fur has three colors—white with orange and black spots, for example—you can almost be certain that the cat is female.

READING CHECK **Apply Concepts** How does a Barr body help explain why the fur of female cats can have three colors?

BUILD VOCABULARY

Academic Words The noun consequence means "a result or an effect of a condition or an action." One consequence of the Y chromosome's having so few genes is that recessive alleles on the X chromosome are expressed in males. A consequence of having two X chromosomes is that females do not show recessive X-linked traits unless they inherit two recessive alleles.

ANIMATION

hhmi | BioInteractive

Figure 15-6

X-Chromosome Inactivation

Calico cats are tricolored. Spots are either orange or black, depending on which X chromosome is inactivated in different patches of their skin.

READING TOOL

As you read about human pedigrees, study the example shown in **Figure 15-7**. Trace the inheritance of the dominant allele.

INTERACTIVITY

Investigate human inheritance with interactive pedigree charts.

Human Pedigrees

Given the complexities of genetics, how would you determine whether a trait is caused by a dominant or recessive allele and whether the gene for that trait is autosomal or sex-linked? The answers, not surprisingly, can be found by applying Mendel's basic principles.

To analyze the pattern of inheritance followed by a particular trait, you can use a **pedigree** chart that shows the relationships within a family. A pedigree shows the presence or absence of a trait according to the relationships among parents, siblings, and offspring.

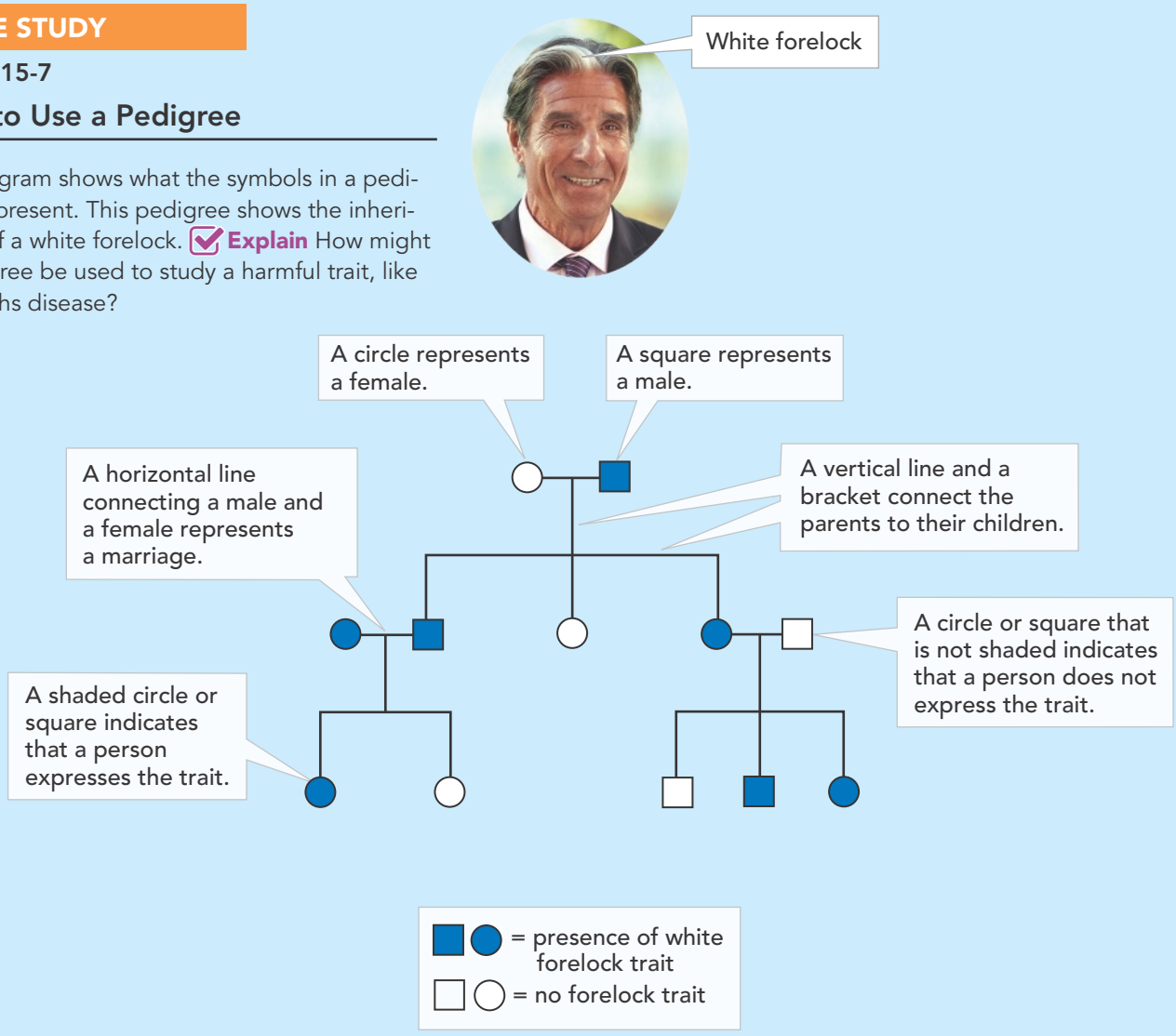
The pedigree in **Figure 15-7** shows how one human trait—a white lock of hair just above the forehead—passes through three generations of a family. The allele for the white forelock trait is dominant. At the top of the chart is a grandfather who had the white forelock trait. Two of his three children inherited the trait. Three grandchildren have the trait, but two do not.

CASE STUDY

Figure 15-7

How to Use a Pedigree

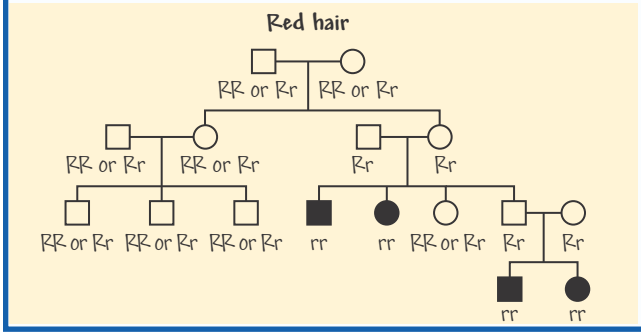
The diagram shows what the symbols in a pedigree represent. This pedigree shows the inheritance of a white forelock. **Explain** How might a pedigree be used to study a harmful trait, like Tay-Sachs disease?



Quick Lab Guided Inquiry

How Can You Analyze a Pedigree?

1. In a small group, choose one of the genetic conditions or disorders described in this chapter.
2. Construct a pedigree for the condition, such as the one for red hair shown here. Begin the pedigree with one pair of grandparents, and then add several children and grandchildren. For each family member, record a genotype for the condition or disorder.



3. Draw a second pedigree, this time showing the phenotype of the condition for each family member.
4. Exchange the second pedigree with another student group.

ANALYZE AND CONCLUDE

1. **Construct an Explanation** Study the pedigree that the other student group produced. How is the genetic condition or disorder inherited? Construct an explanation based on the information the pedigree shows.
2. **Analyze Data** Write a possible genotype for each family member shown in the pedigree.
3. **Evaluate Reasoning** Compare the genotypes you identified with those specified by the student group that produced the pedigree. Explain any differences.

By analyzing a pedigree, we can infer genotypes and predict future outcomes. Because the white forelock trait is dominant, the family members in Figure 15-7 lacking this trait must have homozygous recessive alleles. One of the grandfather's children lacks the white forelock trait, so the grandfather must be heterozygous. On this basis, we can predict that roughly half of the grandfather's children would display the trait.

With pedigree analysis, it is possible to apply the principles of Mendelian genetics to humans. ***The information gained from pedigree analysis makes it possible to determine the nature of genes and alleles associated with inherited human traits.*** Based on a pedigree, you can often determine if an allele for a trait is dominant or recessive, as well as autosomal or sex-linked.

HS-LS3-1

LESSON 15.1 Review

KEY QUESTIONS

1. How is a karyotype made?
2. How are recessive traits inherited when multiple alleles or sex-linked genes are involved?
3. What does a pedigree show about human traits?

CRITICAL THINKING

4. **Construct an Explanation** Why is colorblindness more common in men than in women?
5. **Use Models** A woman with type O blood and a man with type AB blood have children. Use **Figure 15-4** as a model to list the possible genotypes and phenotypes of their children.
6. **Identify Patterns** Is it possible that any of the individuals in the pedigree in **Figure 15-7** are homozygous dominant? Why or why not? What phenotype would you expect to see in the children of an individual who is homozygous dominant for the white forelock trait?
7. **CASE STUDY** Healthy parents may have children who suffer from Tay-Sachs disease, a genetic disorder. Based on this evidence, is Tay-Sachs caused by a dominant or recessive allele? Explain.

LESSON 15.2

Human Genetic Disorders

KEY QUESTIONS

- *What are the effects of errors in meiosis?*
- *How do small changes in DNA affect human traits?*

Normal red blood cells and sickle cells

HS-LS3-1: Ask questions to clarify relationships about the role of DNA and chromosomes in coding the instructions for characteristic traits passed from parents to offspring.

HS-LS3-2: Make and defend a claim based on evidence that inheritable genetic variations may result from: (1) new genetic combinations through meiosis, (2) viable errors occurring during replication, and/or (3) mutations caused by environmental factors.

VOCABULARY

nondisjunction

READING TOOL

As you read, identify the main ideas and details that support the main ideas. Fill in the table in your **Biology Foundations Workbook.**

INTERACTIVITY

Investigate genetic disorders and how they are expressed in humans.

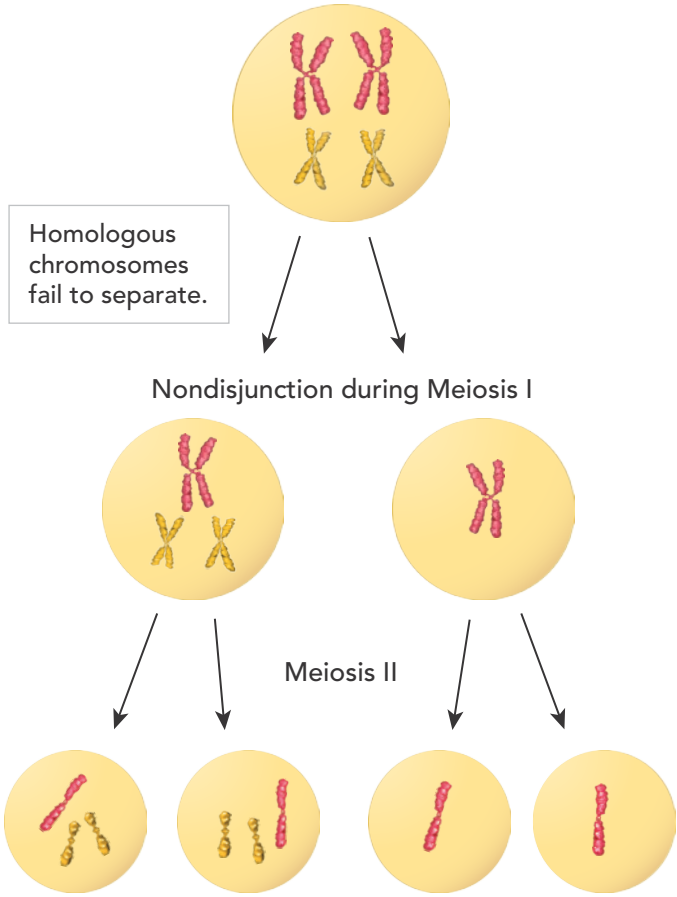
Have you ever heard the expression "It runs in the family"? Relatives or friends might have said that about your smile or the shape of your ears, but what could it mean when they talk of diseases and disorders? What, exactly, is a genetic disorder?

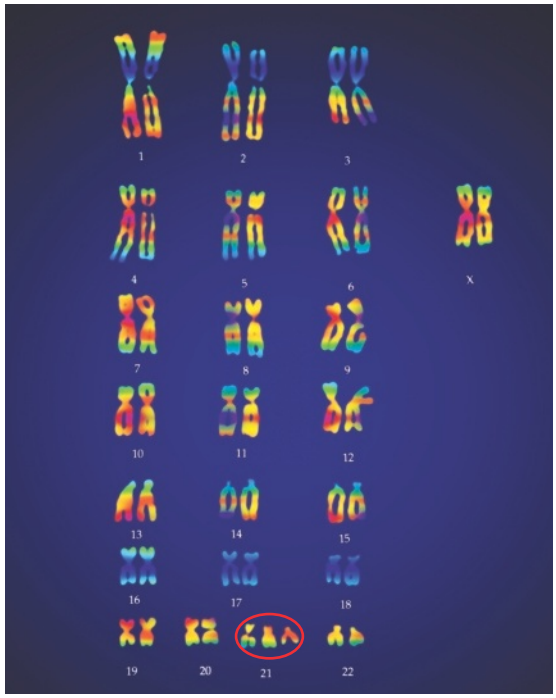
Chromosomal Disorders

Most of the time, the process of meiosis works perfectly and each human gamete gets exactly 23 chromosomes. Every now and then, however, something goes wrong. The most common error in meiosis occurs when homologous chromosomes fail to separate. This mistake is known as **nondisjunction**, which means "not coming apart." **Figure 15-8** illustrates the process.

If nondisjunction occurs during meiosis, gametes with an abnormal number of chromosomes may result, leading to a disorder of chromosome numbers. For example, if two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with three copies of that chromosome. This condition is known as a trisomy, meaning "three bodies." The most common form of trisomy, involving three copies of chromosome 21, is Down syndrome, which is associated with a range of cognitive disabilities and certain birth defects.

Nondisjunction of the X chromosomes can lead to a disorder known as Turner's syndrome. A female with Turner's syndrome usually inherits only one X chromosome. Most women with Turner's syndrome are unable to reproduce because their sex organs do not develop properly at puberty. In males, nondisjunction may cause Klinefelter's syndrome, resulting from the inheritance of an extra X chromosome, which interferes with meiosis and may prevent these individuals from reproducing.





Down Syndrome

Figure 15-8

Nondisjunction

This failure of meiosis causes gametes to have an abnormal number of chromosomes. The karyotype shown here is that of Down syndrome. Notice that there are three copies of chromosome 21 instead of two.

From Molecule to Phenotype

We know that genes are made of DNA and that they interact with the environment to produce an individual organism's characteristics, or phenotype. However, when a gene fails to work or works improperly, serious problems can result.

Molecular research techniques have shown us a direct link between genotype and phenotype. For example, the wax that sometimes builds up in our ear canals can be one of two forms: wet or dry. People of African and European ancestry are more likely to have wet earwax—the dominant form. Those of Asian or Native American ancestry most often have the dry form, which is recessive. The cause is a single DNA base in the gene for a membrane-transport protein. A simple base change from guanine (G) to adenine (A) causes this protein to produce dry earwax instead of wet earwax.

The connection between molecule and trait, and between genotype and phenotype, is often that simple, and just as direct. ***Changes in a gene's DNA sequence can change proteins by altering their amino acid sequences, which may directly affect an individual's phenotype.*** Sometimes, however, these effects are more subtle. For example, certain alleles are associated with tendencies to develop conditions such as diabetes, heart disease, and cancer. Many other factors, such as behavior, diet, and environment, can have a profound effect on whether these conditions actually develop.

BUILD VOCABULARY

Prefixes The word *nondisjunction* is formed by adding the prefixes *non-* and *dis-* to *junction,* which comes from a Latin verb that means "to join." *Non-* (not) and *dis-* (separate) also come from Latin. Put together, these word parts indicate that something joined does not come apart.

Disorders Caused by Individual Genes

Given the size and complexity of the human genome, it shouldn't be surprising that things can sometimes go wrong. In fact, we now know of thousands of genetic disorders caused by changes in individual genes. These changes often affect specific proteins associated with important cellular functions.

READING TOOL

As you read about a disorder, identify the genetic change that causes it.

Sickle Cell Disease

Although sickle cell disease had been known for centuries in Africa, it was not described in the scientific literature until 1910. In the late 1940s, scientists made two important discoveries. The first was that sickle cell disease was hereditary and caused by a recessive allele. The second was that the hemoglobin in people with sickle cell disease was different from normal hemoglobin. Hemoglobin is the oxygen-carrying protein in red blood cells. These two discoveries revealed the links between genes and abnormal proteins, and between proteins and human disease. Sickle cell disease was one of the first recognized molecular diseases, and its discovery spurred research into the molecular origins of other diseases.

As shown in **Figure 15-9**, sickle cell disease is caused by a defective allele for beta-globin, one of two polypeptides in hemoglobin. The defective polypeptide makes hemoglobin a bit less soluble, causing hemoglobin molecules to stick together when the blood's oxygen level decreases. As a result, hemoglobin tends to clump into long fibers that push against the membranes of red blood cells and distort their shape. The physician who first observed such cells described them as "sickle-shaped," and that is how the disorder has been known ever since.

Sickle-shaped cells are more rigid than normal red blood cells, so they tend to get stuck in capillaries—the narrowest blood vessels in the body. If the blood stops moving through the capillaries, damage to cells, tissues, and even organs can result.

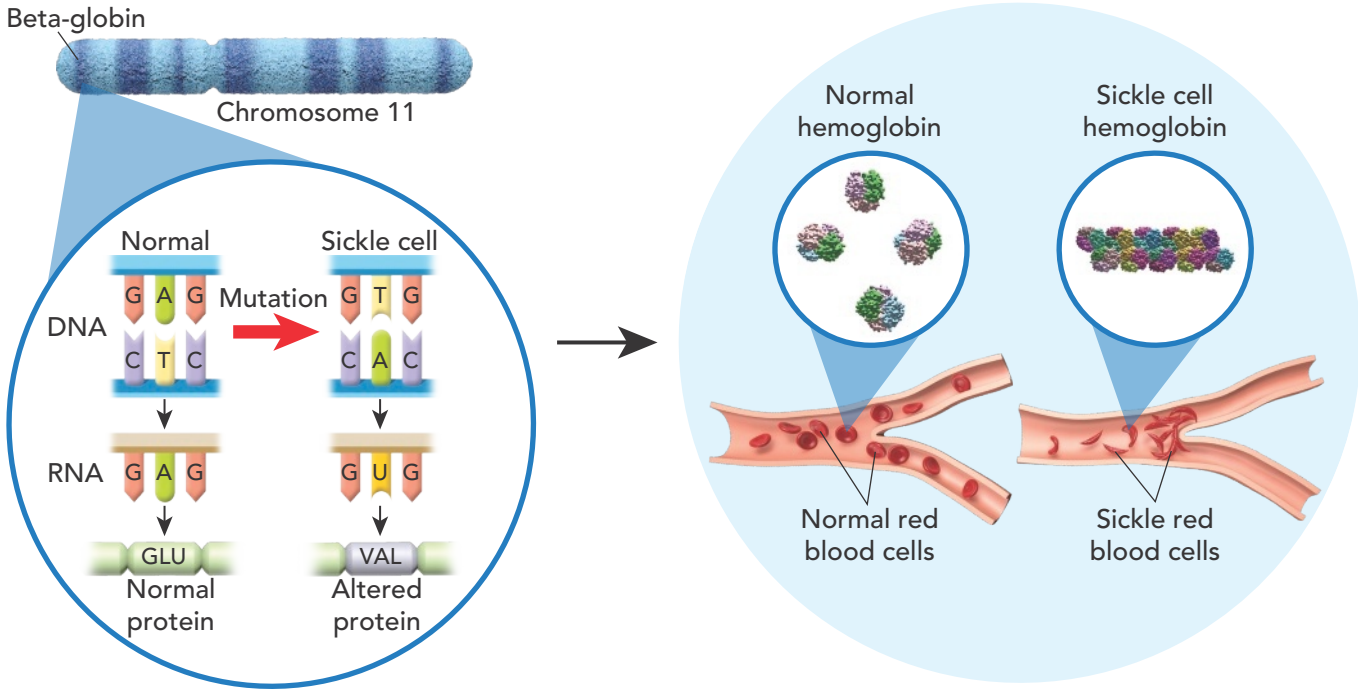
ANIMATION

hhmi | BioInteractive

Figure 15-9

Sickle Cell Disease

A mutation in the beta-globin gene on chromosome 11 results in the formation of aggregated fibers of hemoglobin, which produces the sickle-cell shape of red blood cells.



Cystic Fibrosis Cystic fibrosis (CF) usually results from the deletion of just three bases in the gene for a protein called cystic fibrosis transmembrane conductance regulator (CFTR). CFTR normally allows chloride ions (Cl^-) to pass across cell membranes. The loss of these bases removes a single amino acid—phenylalanine—from CFTR, causing the protein to fold improperly.

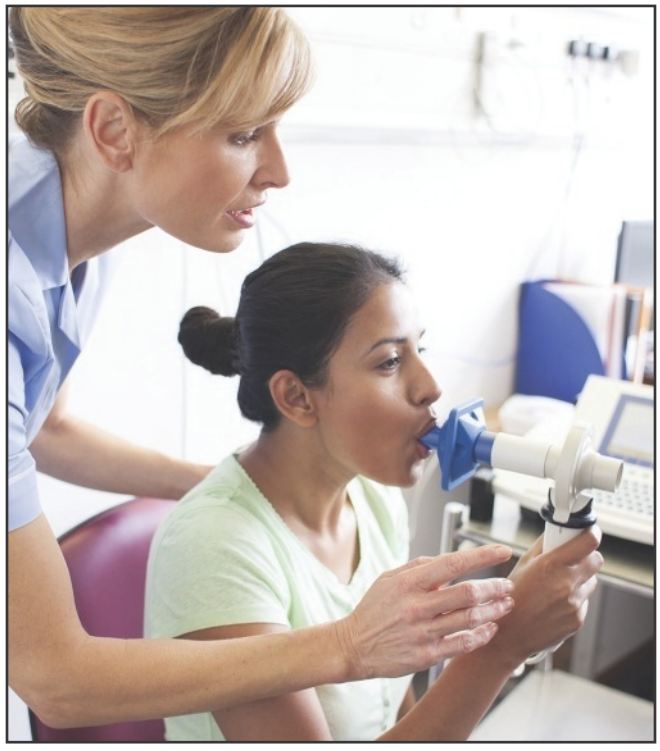
People with one normal copy of the CF allele are unaffected by CF because they can produce enough CFTR to allow their cells to work properly. Two copies of the defective allele are needed to produce the disorder, which means the CF allele is recessive. Children with CF have serious digestive problems and produce thick, heavy mucus that clogs their lungs and breathing passageways. A CF patient undergoing a breathing treatment is shown in **Figure 15-10**.

CASE STUDY

Figure 15-10

CF Treatment

A patient who suffers from cystic fibrosis receives a breathing treatment to help clear the thick mucus in her lungs. **Predict** If both parents are heterozygous for CF, what is the probability that their offspring will suffer from cystic fibrosis?



Huntington's Disease Huntington's disease is caused by a dominant allele for a protein found in brain cells. The allele for this disease contains a long string of bases in which the codon CAG—coding for the amino acid glutamine—repeats over and over again, more than 40 times. Despite intensive study, the reason why these long strings of glutamine cause disease is still not clear. The symptoms of Huntington's disease, namely mental deterioration and uncontrollable movements, usually do not appear until middle age. The greater the number of codon repeats, the earlier the disease appears, and the more severe its symptoms are.

READING CHECK **Infer** Is a person who inherits a single allele for Huntington's disease likely to develop the disorder? If so, when?

Genetic Advantages

Disorders such as CF and sickle cell disease are still common in human populations. In the United States, the CF allele is carried by roughly 1 person in 25 of European ancestry, and approximately 1 person in 12 of African ancestry carries the sickle cell allele. Why are these alleles still around if they can be fatal for those who carry them? The answers may surprise you.

VIDEO

Explore how muscle fibers twitch at different rates and how this affects athletes.

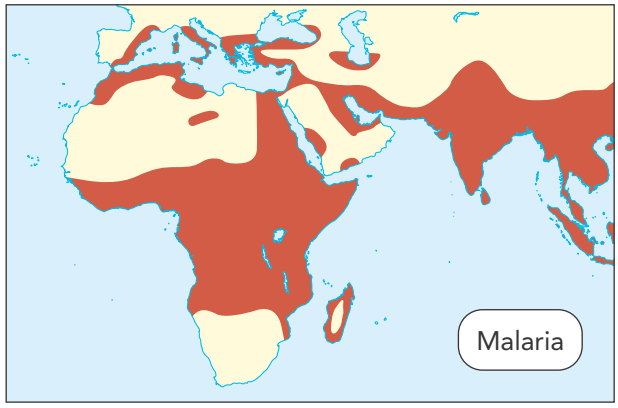
CF Allele and Typhoid More than 1000 years ago, the cities of medieval Europe were ravaged by epidemics of typhoid fever. Typhoid is caused by a bacterium that enters the body through cells in the digestive system. The protein produced by the CF allele helps block the entry of this bacterium. Individuals who are heterozygous for CF would have had an advantage when living in cities with poor sanitation and polluted water. Because they also carried the normal allele, these individuals would not have suffered from cystic fibrosis.

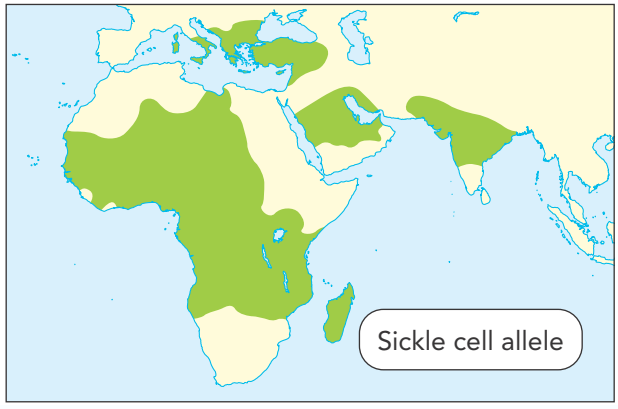
Analyzing Data

The Geography of Malaria

Malaria is a potentially fatal disease transmitted by mosquitoes. Its cause is a parasite that lives inside red blood cells. The upper map shows where malaria is common. The lower map shows regions where the sickle cell allele is the most prevalent.

1. **Analyze Data** What is the relationship between the places where malaria is common and where the sickle cell allele is commonly found?
2. **Construct an Explanation** In 1805, a Scottish explorer named Mungo Park led an expedition of European geographers to find the source of the Niger River in Africa. The journey began with a party of 45 Europeans. During the expedition, most of these men perished from malaria. Their native guides and the other natives they encountered were seemingly unaffected. Use the data to construct an explanation for why the native Africans' phenotype helped them survive.
3. **Form a Hypothesis** As the map shows, the sickle cell allele is not found in African populations that are native to southern Africa. Form a hypothesis to account for this discrepancy.





Sickle Cell Allele and Malaria Many African Americans today are descended from populations that originally lived in west central Africa, where malaria is common. Malaria is a mosquito-borne infection caused by a parasite that lives inside red blood cells. Individuals with just one copy of the sickle cell allele are generally healthy and are also highly resistant to the parasite. This resistance gives them a great advantage against malaria, which even today claims more than four hundred thousand lives every year.

HS-LS3-1, HS-LS3-2

LESSON 15.2 Review

KEY QUESTIONS

1. How does nondisjunction affect the production of gametes?
2. How can a change in genotype affect phenotype?

CRITICAL THINKING

3. **Use Models** What condition is modeled by the expression 47, XXY? Explain what the model shows.
4. **Synthesize Information** Is it possible for a change of a single nucleotide in DNA to have an observable effect on phenotype? Include an example to support your answer.
5. **Construct an Explanation** How can alleles that cause serious diseases, such as sickle cell disease, still provide an advantage to the human population?

Studying the Human Genome

LESSON 15.3

KEY QUESTIONS

- *How can scientists read DNA base sequences?*
- *What research efforts have resulted from the Human Genome Project?*

HS-LS3-1: Ask questions to clarify relationships about the role of DNA and chromosomes in coding the instructions for characteristic traits passed from parents to offspring.

HS-ETS1-1: Analyze a major global challenge to specify qualitative and quantitative criteria and constraints for solutions that account for societal needs and wants.

There are more than 6 billion base pairs of DNA in just a single human cell. If we think of each base pair as a single letter, that amounts to nearly 3000 textbooks of 1000 pages each. Just a few decades ago, that might have seemed to be an unimaginably large amount of information. Today, however, we have powerful techniques to read, store, and process that information. As a result, we know more about the human genome than ever—and are learning more every day.

Manipulating DNA

Since the discovery of the double helix, biologists have dreamed of a time when they could read the DNA sequences in the human genome. For a long time, it seemed impossible. DNA molecules are huge—even the smallest human chromosome contains nearly 50 million base pairs. Manipulating such large molecules is difficult. In the late 1960s, however, scientists discovered natural enzymes, called **restriction enzymes**, that could cut DNA at specific sites. **Figure 15-11** shows how restriction enzymes work. ***By using tools that cut, separate, and copy nucleic acids, scientists can now read DNA base sequences.*** Such techniques have made it possible to study the genomes of living organisms, including humans, in great detail.

VOCABULARY

restriction enzyme
gel electrophoresis
genomic imprinting

READING TOOL

As you read, keep track of the different steps that scientists use to read nucleotide sequences. Fill in the graphic organizer in your **Biology Foundations Workbook.**

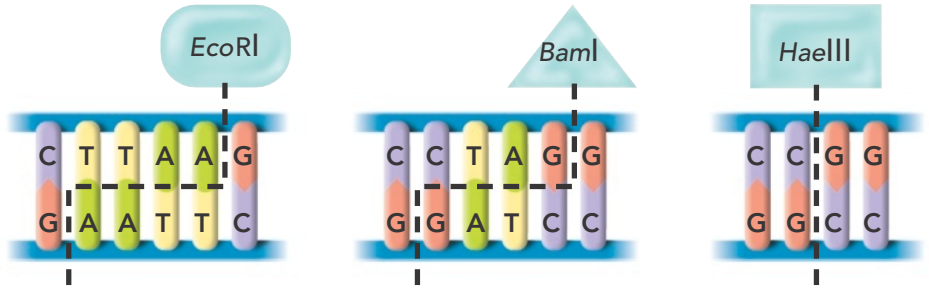
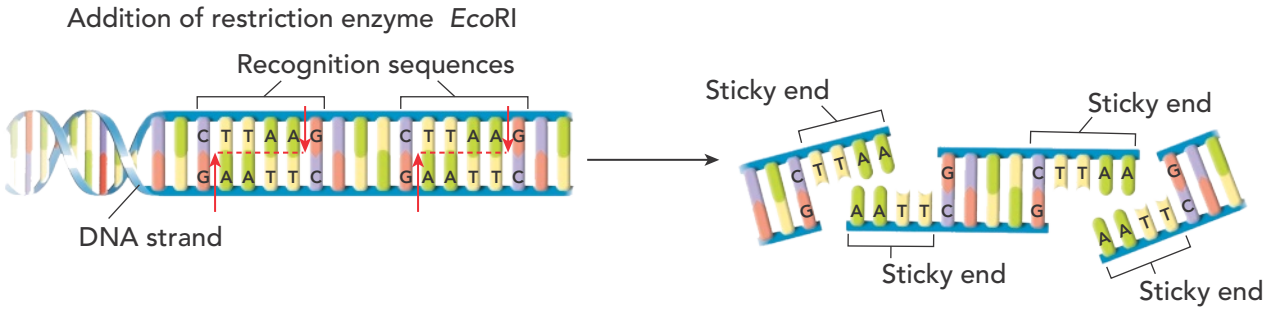


Figure 15-11
Restriction Enzymes

These enzymes recognize specific DNA sequences, and then cut both DNA strands as shown.



INTERACTIVITY

Figure 15-12

Cutting DNA

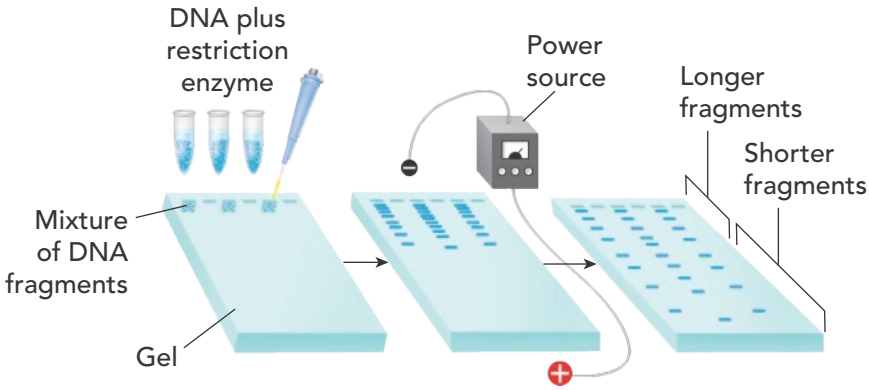
Restriction enzymes cut DNA in specific ways.

Cutting DNA DNA is relatively easy to extract from cells and tissues. However, DNA molecules are so large that they must first be cut into smaller pieces for analysis. Many bacteria produce restriction enzymes that do exactly that. They precisely cut DNA molecules into smaller pieces, several hundred bases in length. Bacteria use restriction enzymes to cut and inactivate the DNA of bacteriophages, viruses that infect bacteria. A restriction enzyme is like a key that fits only one lock. For example, as shown **Figure 15-12**, the *Eco*RI restriction enzyme can only recognize the base sequence GAATTC. It cuts each strand of DNA between the G and A bases, leaving single-stranded overhangs with the sequence AATT. The overhangs are called "sticky ends" because they can bond, or "stick," to a DNA fragment with the complementary base sequence.

Figure 15-13

Separating DNA

An electric voltage moves DNA fragments across a gel similar to a slice of gelatin. Within an hour or two, the fragments all separate, each appearing as a band on the gel.



INTERACTIVITY

Conduct a virtual activity using restriction enzymes to isolate particular genes.

Separating DNA Once DNA has been cut by restriction enzymes, scientists can use a technique known as **gel electrophoresis** to separate the fragments. **Figure 15-13** illustrates this simple, yet effective, method. A mixture of DNA fragments are placed in wells on one end of a porous gel. When an electric voltage is applied to the gel, DNA molecules—which are negatively charged—move toward the positive end of the gel. The smaller the DNA fragment, the faster and farther it moves. The current is turned off after a few hours. The result is a pattern of bands based on fragment size. Specific stains that bind to DNA make these bands visible. Researchers can then remove individual restriction fragments from the gel and study them further.

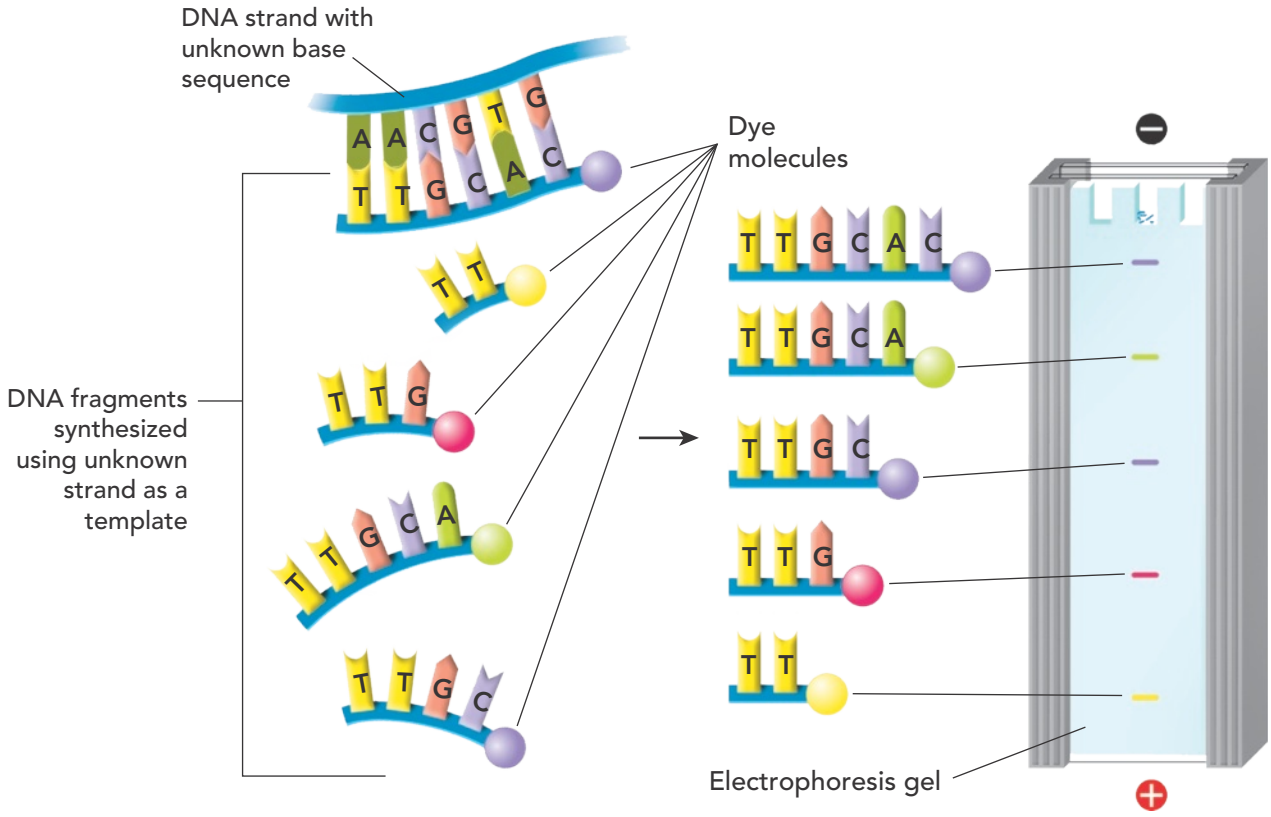


Figure 15-14
Reading DNA

Fragments of DNA separated by electrophoresis and tagged with dye can be used to determine the sequence of bases in a strand of DNA.

Reading DNA After the DNA fragments have been separated, researchers use a clever chemical "trick" to read, or sequence, them. The single-stranded DNA fragments are placed in a test tube containing DNA polymerase—the enzyme that copies DNA—along with the four nucleotide bases, A, T, G, and C. As the enzyme goes to work, it uses the unknown strand as a template to make one new DNA strand after another. The tricky part is that researchers also add a small number of bases that have a chemical dye attached. Each time a dye-labeled base is added to a new DNA strand, the synthesis of that strand stops. When DNA synthesis is completed, the result is a series of color-coded DNA fragments of different lengths, as shown in **Figure 15-14**. Researchers can then separate these fragments, often by gel electrophoresis. The order of colored bands on the gel tells the exact sequence of bases in the DNA.

Until the late 1980s, the sequencing of DNA was done by researchers painstakingly analyzing the columns and bands on electrophoresis gels and recording their data. This was a slow process, prone to errors, and very expensive. It takes a person about a year to read a sequence of 20,000 to 50,000 bases—and the human genome contains about 3 billion base pairs. Fortunately, sequencing has been improved and automated over the decades, becoming faster, less expensive, and more accurate. Tasks that once took a person a year to do can now be done in a matter of hours. Some labs now sequence well over 100,000 billion bases per year, with hundreds of DNA fragments being sequenced simultaneously.

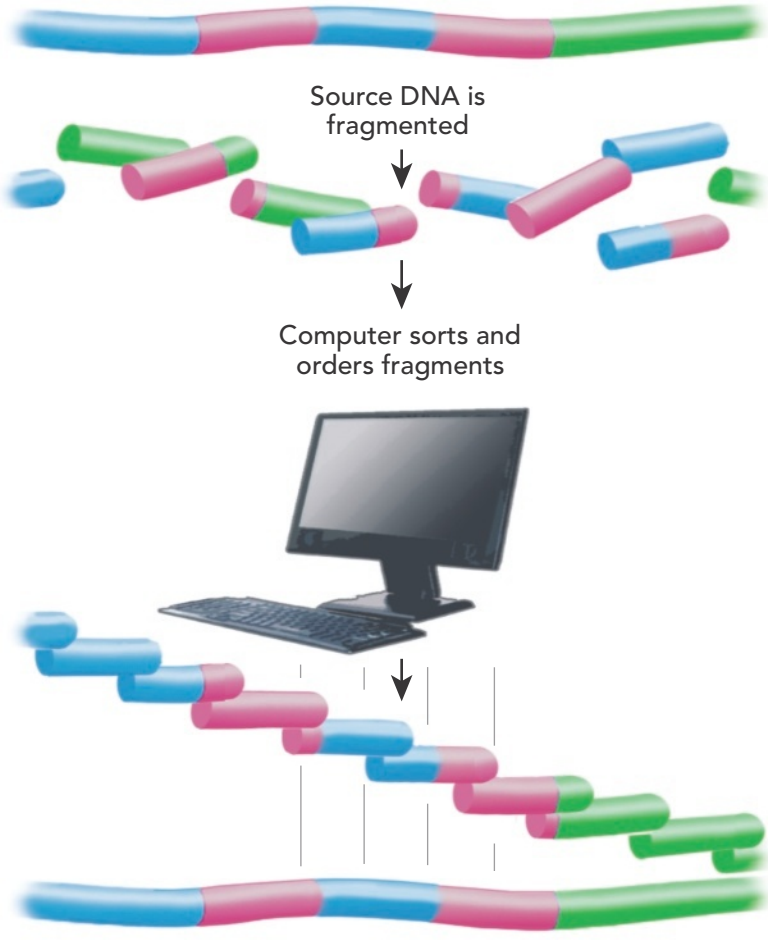
INTERACTIVITY

Investigate how genomic sequencing is performed using restriction enzymes and fragments of DNA.

READING TOOL

After you have read this section, write down the sequence of events involved in manipulating and reading DNA. Reread the section to check your work.

Assembling the Sequence One common feature of most DNA sequencing techniques is that they read fragments no more than a few hundred bases in length. How are these short "reads" of DNA put together to produce a full-length map? **Figure 15-15** shows how this is done through a technique known as "shotgun" sequencing. Whole chromosomes are cut into random fragments. These are sequenced automatically, and the information is fed into a computer. A computer program analyzes the data by searching for matching sequences among the fragments, and aligns them to reassemble the fragments and complete the sequence.



Overlapping sequences are matched and aligned to determine the complete DNA sequence.

Figure 15-15

Putting the Pieces Together

Computers can quickly find overlapping sequences, thereby determining the sequence for an entire length of DNA.

Some new sequencing technologies have even done away with the need to cut DNA into fragments. One involves drawing a strand of DNA through a tiny pore, like a thread being pulled through the eye of a needle. Each base creates a different kind of electrical signal as it passes through the pore, allowing the bases to be read one by one. As a result of all this technical progress, you might say that the "easy" part of genome analysis today is reading the DNA base sequence. The hard part is making sense of it.

READING CHECK **Summarize** Describe how DNA is sequenced.

HS-LS3-1

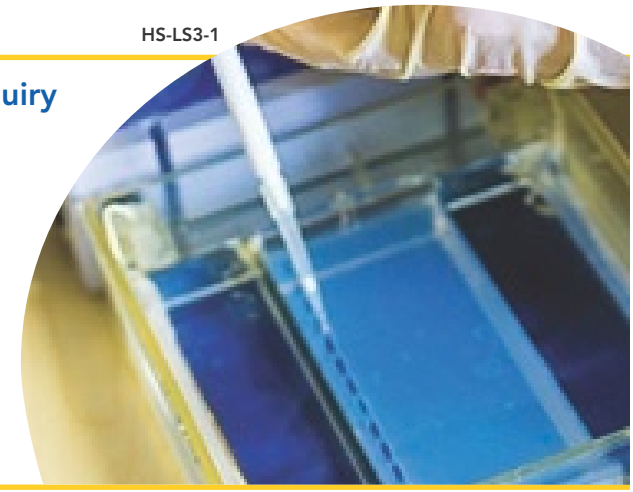
Develop a Solution Lab — Guided Inquiry

Gel Electrophoresis

Problem How can molecules be separated by electric charge?

Scientists use gel electrophoresis to separate DNA fragments. In this lab, you will practice the technique of gel electrophoresis by separating food dye into its different components.

You can find this lab in your digital course.



The Human Genome—What's Inside?

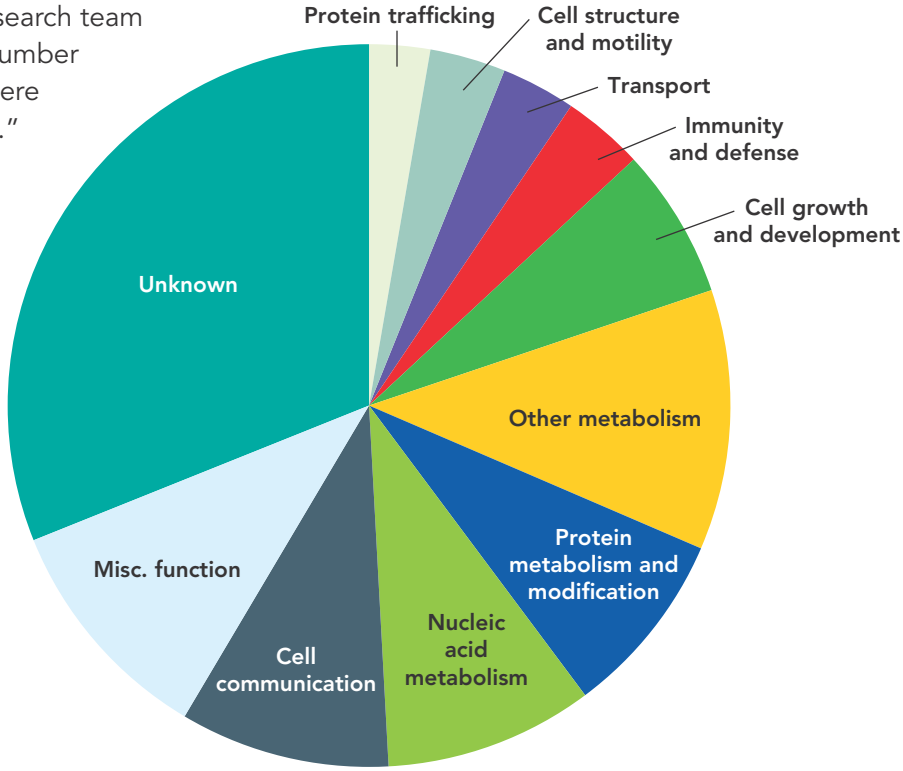
In 2003, an international effort known as the Human Genome Project finished the first complete human DNA sequence. But scientists quickly realized there was much more to be done, and set about trying to make sense of all the data. ***Labs around the world now study which regions of DNA are transcribed into RNA, which bind to proteins, which are marked with epigenetic tags, and which vary from one individual to the next.*** The Human Genome Project was only the beginning of a new era of research on human molecular genetics, but the findings so far are fascinating.

How Many Genes? Research on the human genome revealed many surprises. Human cells contain approximately 20,000 genes, which is not a large number compared to the cells of other species. In fact, one research team wrote that with respect to the number of genes, humans are "somewhere between a chicken and a grape." The chicken genome has about 17,000 genes, and the grape has about 30,000 genes. What do those genes code for? As **Figure 15-16** shows, the functions of more than a quarter of human genes are unknown. The other genes fall into categories such as transcription factors, metabolic enzymes, components of the cell membrane, receptors, and regulatory factors.

Figure 15-16

Functions of Human Genes

While the functions of many genes are known, we are still unsure of the functions of more than 25 percent of our genes.



Adapted from Freeman Biological Science ©2011

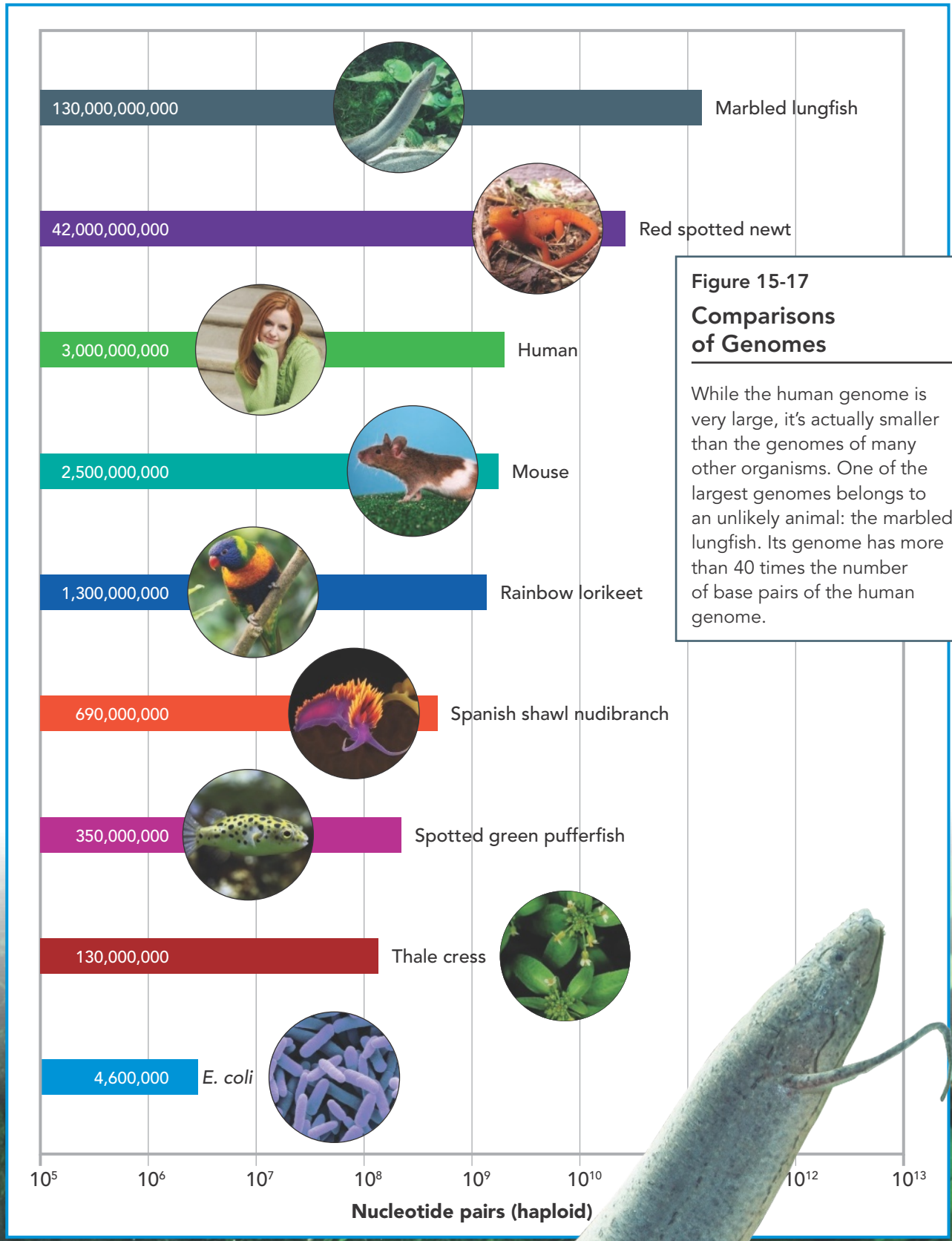


Figure 15-17

Comparisons of Genomes

While the human genome is very large, it's actually smaller than the genomes of many other organisms. One of the largest genomes belongs to an unlikely animal: the marbled lungfish. Its genome has more than 40 times the number of base pairs of the human genome.

The Large and Small of It At 3 billion bases, the human haploid genome is larger than the genome of many other organisms. You might be tempted to think that our unique capabilities are the result of having so much DNA. However, the cells of many organisms contain far more DNA than our cells do! The ordinary onion has nearly five times as much DNA as we have. The marbled lungfish (*Protopterus aethiopicus*) has more than 40 times as much. Curiously, another type of fish, the puffer fish (*Fugu*), has the smallest vertebrate genome discovered so far—just 350 million bases. **Figure 15-17** provides a comparison of the genome sizes of many different types of organisms.

One of the most striking aspects of the human genome is how little of it actually codes for proteins—only about 2 percent. So, what does the rest of the DNA code for? Some of it, of course, is involved in the regulation of gene expression. DNA sequences known as enhancers can bind proteins to open up genes for transcription. Other regions are involved in blocking the expression of nearby genes. Many other regions are transcribed at low frequencies, producing RNA molecules that are not translated into proteins but may play a role in gene regulation. However, all of these sequences taken together account for only about 10 percent of the genome.

Approximately 50 percent of the human genome is composed of highly repetitive DNA sequences—short stretches where the base sequence is repeated over and over again. Many, if not most, of these regions are derived from transposable elements, pieces of DNA that move from place to place in the genome and sometimes generate multiple copies of themselves. The functions, if any, of these regions, which make up more than half of the human genome, remain unknown.

READING CHECK **Infer** Is there a relationship between the complexity of an organism and the number of bases in its genome?

The Personal Genome If you were to compare the genomes of two unrelated individuals, you would find that most—but not all—of their DNA will match base for base with each other. On average, about one base in 1200 will not match between two individuals. Biologists call these single base differences single nucleotide polymorphisms (SNPs, or "snips"). Researchers have discovered that certain sets of closely linked SNPs occur together time and time again. Some of these are associated with certain traits, including the susceptibility to particular diseases or medical conditions. Increasingly, high-speed DNA sequencing is making it possible to rapidly pinpoint SNPs and their associated alleles, enabling physicians to tailor medical treatments to a patient's genome. In addition, many private companies now offer a "personal genome" service that analyzes one's DNA for a modest price, and uses that information to determine ethnic ancestry as well as the possibility of developing certain diseases. As we further explore the human genome, the medical and social possibilities for the use of personal genomic information will only increase.

Figure 15-18

Collecting Genetic Information

People who are interested in learning about their ancestry or risk for certain diseases, might collect and send their DNA to private companies for analysis. Many ethical questions and decisions surround this practice.

Genome Privacy Rapid advances in gathering and analyzing genomic data have raised a number of ethical and legal questions. For example, who owns and controls the genetic information of the person in **Figure 15-18**? Who should have access to personal genetic information? In response to some of these issues, in 2008, the U.S. Congress passed the Genetic Information Nondiscrimination Act. This act makes it illegal for insurance companies and employers to discriminate based on information from genetic tests. As the science advances, other protective laws may soon follow.

BUILD VOCABULARY

Use Prior Knowledge You know that a genome refers to the genetic information carried in the DNA of an organism. An imprint is an identifying marker. Therefore, the term *genomic imprinting*, refers to marks that are carried on DNA.

Gene Imprinting As you read in Chapter 14, epigenetic chemical marks can be attached to DNA and histone proteins in a way that affects gene expression by altering chromatin structure. This process is known as **genomic imprinting**. In humans as well as other mammals, some of these marks can be passed from one generation to the next through either the mother or the father. This means there are some genes that are only expressed if they came from a male parent and there are some genes that are only expressed if they came from a female parent. Nearly 100 genes in humans are imprinted in this way.

One example of genomic imprinting is a condition called Angelman syndrome, which results from the deletion of a gene known as UBE3A on chromosome 15. Angelman syndrome causes seizures and an unusually happy disposition. An affected child typically inherits two copies of chromosome 15. One copy comes from the child's mother and one from the child's father.

READING CHECK Review How does genomic imprinting lead to the expression of one allele over another?

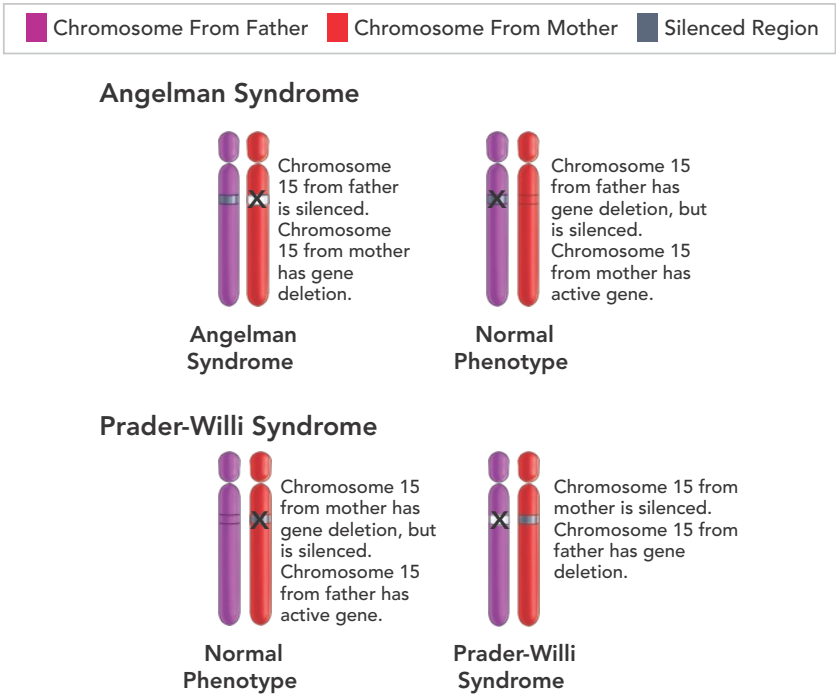


Figure 15-19

Deletions and Imprinting

Whether a child has Angelman syndrome or Prader-Willi syndrome depends on whether the defective chromosome is inherited from the mother or the father.

The region containing this gene is silenced by epigenetic imprinting in the chromosome inherited from the child's father, while the same region inherited from the mother is active. This means that if a child inherits the deletion on chromosome 15 from its mother, the child will develop Angelman syndrome, since the paternal copy of the gene is silenced, as shown in **Figure 15-19**. On the other hand, if a child inherits the deletion from its father, it will have no effect, since that region of the chromosome is silenced by imprinting, and the mother's copy of the gene is active. Interestingly, there is another genetic disorder known as Prader-Willi syndrome caused by the deletion of a nearby region of the same chromosome. This deletion mutation, however, is inherited from the paternal chromosome, because in this region it is the mother's genes that are silenced by genomic imprinting. Prader-Willi syndrome causes weak muscles and feelings of extreme hunger.

HS-LS3-1, HS-ETS1-1

LESSON 15.3 Review

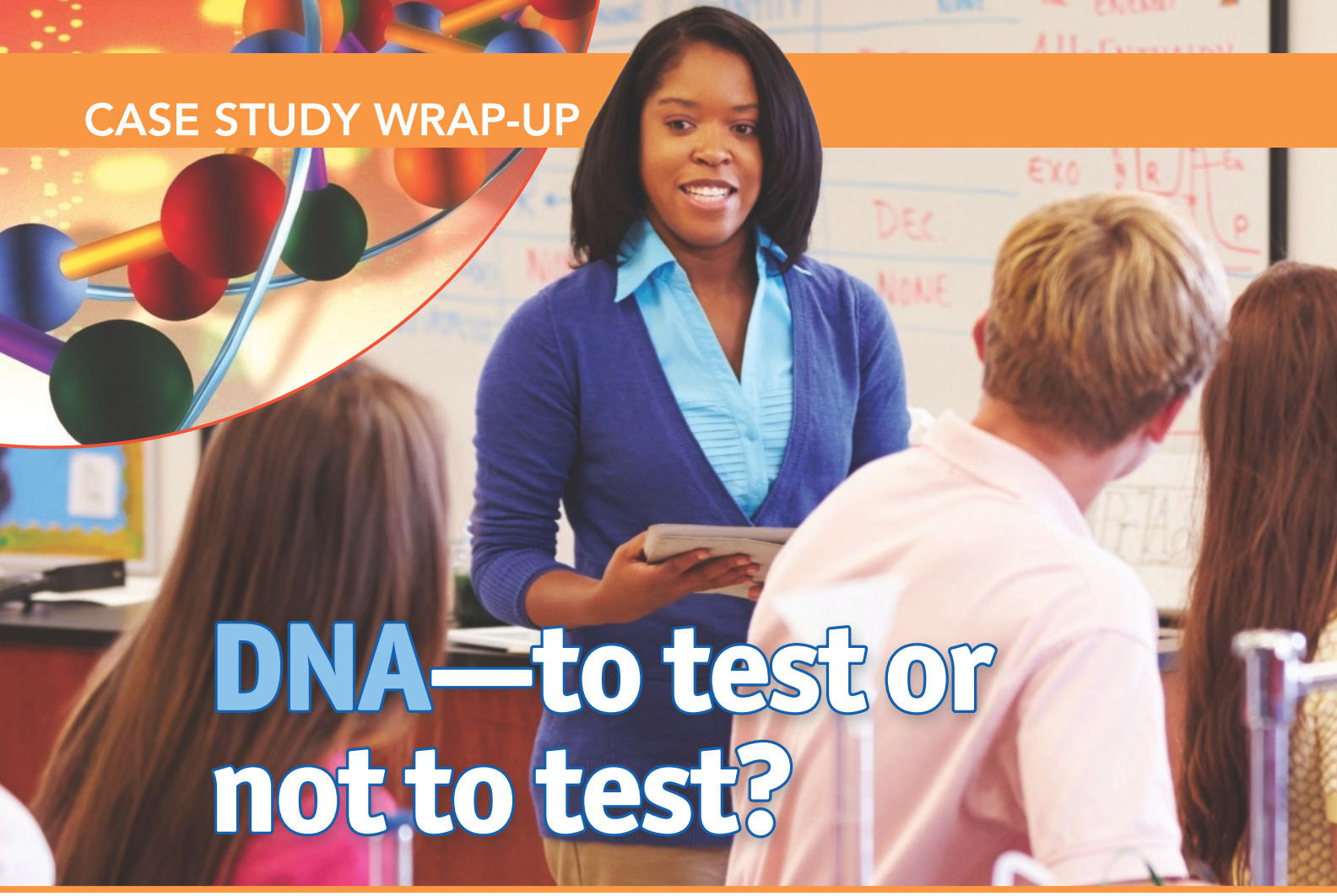
KEY QUESTIONS

1. How are scientists able to read DNA base sequences?
2. How does the number of genes in human cells compare with the numbers in other species?

CRITICAL THINKING

3. **Construct an Explanation** How does gel electrophoresis work to separate DNA fragments?
4. **Apply Scientific Reasoning** A point mutation changes one nucleotide at a random location in human DNA. How likely is the mutation to cause a change in the amino acid sequence of a protein? Apply scientific reasoning to explain your prediction.
5. **CASE STUDY** A couple receives DNA analysis, which determines that they are both heterozygous for the gene responsible for Tay-Sachs disease. Should doctors or the government have a role in deciding if the couple can have children together? Construct an argument to explain why or why not.

CASE STUDY WRAP-UP



DNA—to test or not to test?

Scientists now have the knowledge and tools to analyze DNA. Human relationships can be traced, crimes can be solved, and genetic diseases can be identified. However, with this knowledge comes some serious ethical and legal questions.

HS-LS3-1, HS-ETS1-2, CCSS.ELA-LITERACY.WHST.9-10.2

Make Your Case

DNA testing can identify carriers of the Tay-Sachs allele. In an effort to eliminate this tragic disease, couples at risk are urged to get tested for the allele before they marry or have children. Potentially, this approach could also reduce the incidence of other genetic diseases and disorders.

Develop a Solution

1. **Ask Questions** With a partner, research two common human genetic disorders. Find out whether genetic testing is available for them, and find out whether such tests are generally offered to prospective parents.
2. **Form an Opinion** Decide whether such testing makes medical and economic sense for the disorders you have researched. Discuss whether there should be legal requirements for such testing and what the rights of prospective parents should be in cases where disease-causing alleles are found.

Society on the Case

Private Information?

For many years, people have been volunteering their DNA for research studies. Analyses of the DNA have been published online or in other forums, but never with the name of the volunteer included. For years, scientists assumed that they were safeguarding people's privacy. Now, new knowledge and technology are challenging this assumption.

From analyzing a DNA sample and other genetic information, scientists can now infer data about a person's weight, age, and general health. Some viral infections can be detected because they change genetic activity. Genes also show risks for Alzheimer's disease, cancer, and other diseases.

Is it possible to identify someone from only a DNA sample? In 2013, Yaniv Erlich of the Whitehead Institute in Massachusetts showed a surprising answer to this question. He selected five samples of DNA at random from a database. Using public records and a computational model he developed, he identified each of the five people by name.

Not all DNA samples are volunteered. The U.S. military catalogs the DNA of soldiers. Police officers gather DNA samples from crime scenes, and they attempt to match the samples to criminal suspects. Suspects may be forced to submit a DNA sample.

Should people have the right to keep their DNA private? Or is DNA no more private than a face or fingerprint? What do you think?

Careers on the Case

Work Toward a Solution

Analyzing DNA involves tracking millions, if not billions, of individual nucleotides. Computers make this kind of analysis possible, so computer specialists are essential for DNA research.

Computational Biologist

Most scientists work with computers to organize data. For complex data analysis, however, they might consult a computational biologist. This field applies computer science and math to help solve problems in biology or biological research.

VIDEO

Learn more about careers that combine computers and biology.

Lesson Review

Go to your Biology Foundations Workbook for longer versions of these lesson summaries.

15.1 Human Chromosomes

A genome is the full set of genetic information that an organism carries in its DNA. Scientists arrange images of chromosomes in a genome to produce a karyotype, which shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size. The full complement of chromosomes in the human cell is 46—two sets of 23.

Human traits are passed on largely through dominant and recessive alleles, although some alleles display codominant inheritance. Genes located on sex chromosomes are called sex-linked genes. A pedigree chart traces the presence and absence of traits through generations of a family, and can often determine if an allele for a trait is dominant or recessive, autosomal or sex-linked.

- genome
- karyotype
- sex chromosome
- autosome
- sex-linked gene
- pedigree

Blood Groups		
Phenotype (Blood Type)	**Genotype**	**Antigen on Red Blood Cell**
A	I^AI^A or I^Ai	1.
B	2.	B
3.	4.	A and B
5.	*ii*	6.

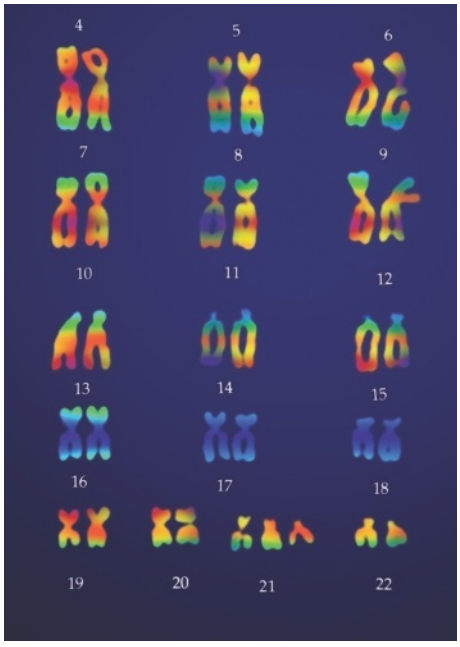
Review Complete the table of human blood types. Which blood types are produced by two different genotypes?

15.2 Human Genetic Disorders

Chromosomal disorders such as nondisjunction in meiosis can produce Down syndrome, which is associated with cognitive disabilities and birth defects, or Turner's and Klinefelter's syndromes, which impair reproductive capacity.

The DNA in genes interacts with the environment to produce an individual phenotype. Changes in a gene's DNA sequence can change proteins by altering their amino acid sequences. These changes can affect an individual's phenotype or, more subtly, underlie tendencies to develop conditions such as diabetes, heart disease, and cancer. These conditions may or may not develop depending on many other factors, such as diet, behavior, and environment. Some changes in specific genes cause disorders such as sickle cell disease, cystic fibrosis, and Huntington's disease.

- nondisjunction



Interpret Data Which chromosome illustrates nondisjunction? How do you know?

15.3 Studying the Human Genome

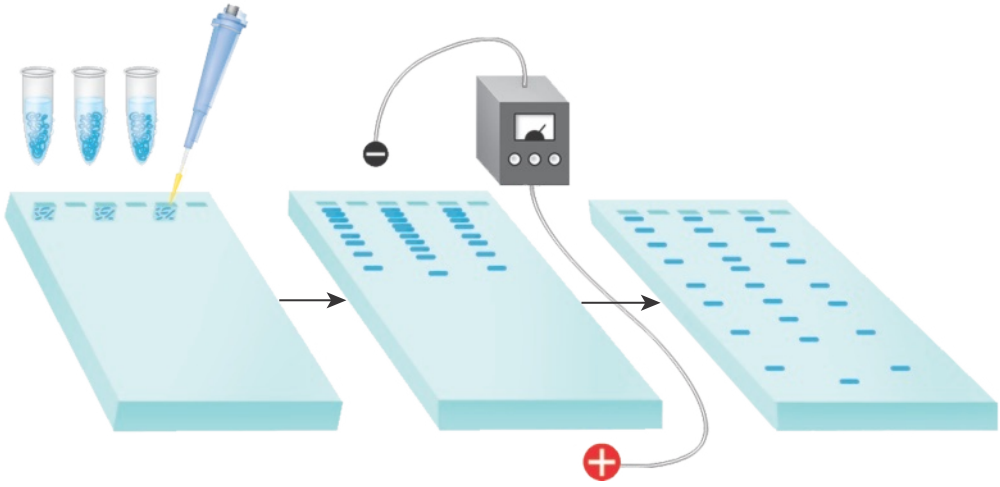
Scientists can use tools that cut, separate, and copy nucleic acids. These tools make it possible to study the genomes of different organisms. Restriction enzymes can be used to cut DNA molecules into smaller pieces. These DNA fragments are then separated using gel electrophoresis. The separated fragments are finally sequenced using a technique that includes copying the DNA. New sequencing technologies have allowed scientists to read DNA without cutting it into fragments.

In 2003, the Human Genome Project finished the first complete DNA sequence. Surprisingly, human cells contain approximately 20,000 genes, which is not a large number compared to other species.

Single nucleotide polymorphisms, or SNPs, are single-base base differences that are found among individuals. Some SNPs are associated with certain traits. Many private companies offer services that analyze one's DNA to determine ethnic ancestry and the possibility of developing certain diseases.

In a process known as genomic imprinting, epigenetic chemical marks on DNA can be passed from one generation to the next. These marks can affect gene expression, and some genes are only expressed if they come from a male parent while others are only expressed if they come from a female parent.

- restriction enzyme
- gel electrophoresis
- genomic imprinting



Interpret Diagrams How does gel electrophoresis separate DNA fragments?

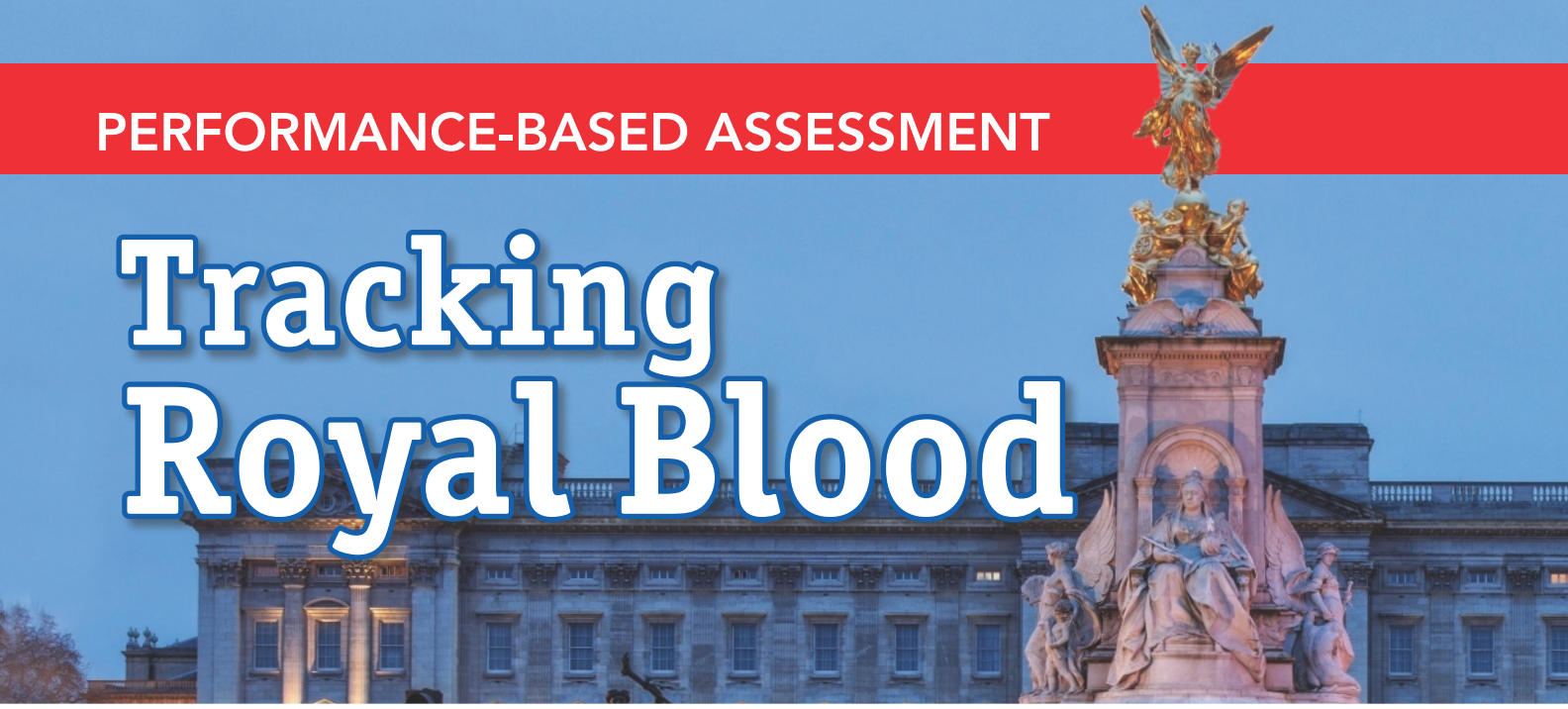
Organize Information

Complete the table with definitions of the terms and explanations of how the concepts help you understand human heredity.

Genetic term	Definition	How concept helps you understand human heredity
genome	1.	2.
karyotype	3.	4.
autosome	5.	6.
nondisjunction	7.	8.
genomic imprinting	9.	10.

PERFORMANCE-BASED ASSESSMENT

Tracking Royal Blood



Evaluate and Communicate Information

HS-LS3-1, CCSS.ELA-LITERACY.WHST.9-10.9

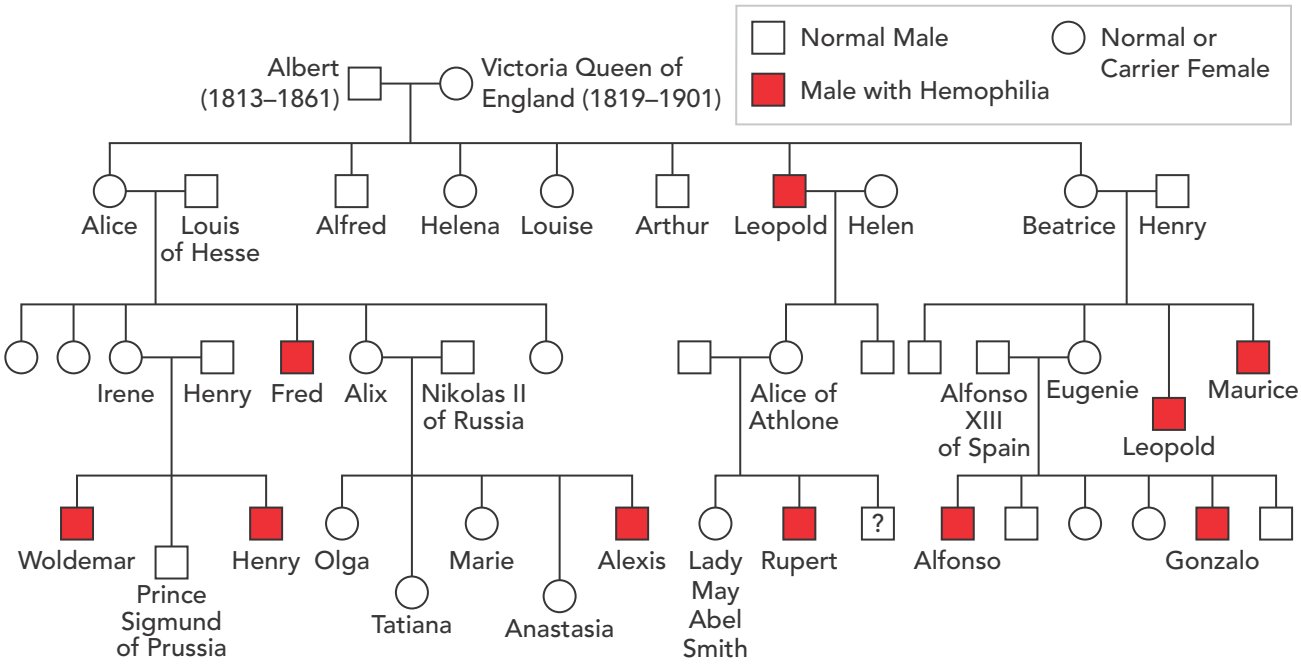
STEM From 1837 until her death in 1901, Queen Victoria ruled Great Britain and the lands it conquered. The queen and her family had a combination of wealth, power, and privilege that might be difficult to imagine today. Nevertheless, their lives were hardly ideal.

The queen's youngest son, Prince Leopold, was weak and sickly. So were some of the queen's grandchildren. They suffered from a disease called hemophilia, in which the blood clots poorly. With this disease, any minor injury can lead to severe bleeding or bruising. In the 1800s, few hemophiliacs lived long into adulthood. Prince Leopold died at age 30.

Today, scientists recognize hemophilia as a genetic disorder. They have identified two types of hemophilia, the specific faulty genes associated with hemophilia, and the mutations that affected them. The queen and her family, however, had no such knowledge. All they could do was recognize the pattern of inheritance and worry about the next child to be born.

Study the pedigree on the next page, which shows some of the queen's family. Notice that females could be carriers. Carriers are individuals who carry the defective gene but do not suffer from the disease. The queen and many of her descendants were carriers.

1. **Analyze Data** Identify the carriers of the gene associated with hemophilia. Explain your reasoning.
2. **Cite Evidence** Is the gene for hemophilia located on an autosome, the X chromosome, or the Y chromosome? Cite the evidence in the pedigree to support your answer.
3. **Draw Conclusions** Is the allele for hemophilia dominant or recessive? How do you know?
4. **Construct an Explanation** Using the family in the pedigree as an example, explain how the trait of hemophilia is expressed, and how it passes through generations.
5. **Construct a Simulation** Work with a partner to construct another pedigree that shows the inheritance of hemophilia, sickle cell disease, or another genetic disorder or condition presented in this chapter. Draw the pedigree and present it on a poster, or prepare a computer presentation. Share your pedigree with the class, and explain the pattern of inheritance that it shows.


Normal Male
Male with Hemophilia
Normal or Carrier Female
Albert (1813–1861)
Victoria Queen of England (1819–1901)
Alice
Louis of Hesse
Alfred
Helena
Louise
Arthur
Leopold
Helen
Beatrice
Henry
Irene
Henry
Fred
Alix
Nikolas II of Russia
Alice of Athlone
Alfonso XIII of Spain
Eugenie
Leopold
Maurice
Woldemar
Prince Sigmund of Prussia
Henry
Olga
Tatiana
Marie
Anastasia
Alexis
Lady May Abel Smith
Rupert
?
Alfonso
Gonzalo

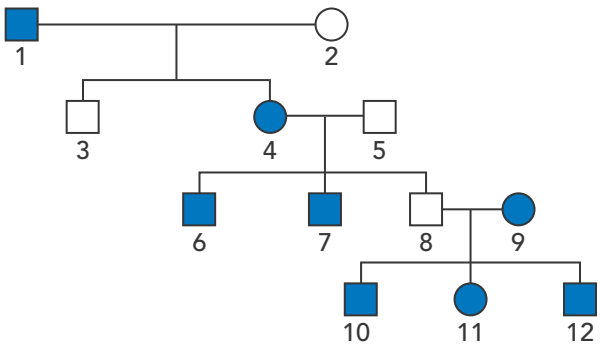
ASSESSMENT

KEY QUESTIONS AND TERMS

15.1 Human Chromosomes

HS-LS3-1, HS-LS3-2

1. What is the number of chromosomes in a normal human karyotype?
   a. 2 **c**. 44
   b. 23 **d**. 46
2. Colorblindness is more common in males than in females because the allele for colorblindness is
   a. dominant and located on the X chromosome.
   b. dominant and located on the Y chromosome.
   c. recessive and located on the X chromosome.
   d. recessive and located on the Y chromosome.
3. The alleles for blood groups I^A and I^B are codominant. When paired they produce the blood type
   a. A. **c**. O.
   b. B. **d**. AB.
4. Explain why there are only 23 chromosomes in a sperm cell or an egg cell.
5. Why is blood type O considered the universal donor?
6. What phenomenon occurs in females as a result of X-chromosome inactivation?
7. How does a pedigree chart differ from a family tree?
8. How many generations are shown in this diagram?



15.2 Human Genetic Disorders

HS-LS3-1

9. Which of the following diseases and conditions does not appear until later in a person's life?
   a. cystic fibrosis
   b. sickle cell disease
   c. colorblindness
   d. Huntington's disease
10. Because two copies of a defective CF allele are needed to produce cystic fibrosis,
    a. the CF allele is dominant.
    b. the CF allele is recessive.
    c. the CF allele cannot produce CFTR.
    d. the CF allele overproduces CFTR.
11. Which of the following disorders does NOT result from nondisjunction in meiosis?
    a. Down syndrome
    b. Turner's syndrome
    c. Klinefelter's syndrome
    d. sickle cell disease
12. How can being heterozygous for sickle cell disease be beneficial rather than harmful?
13. What function of the CF allele protects a carrier from contracting typhoid?
14. Nondisjunction of the X chromosomes produces what disorder in women?
15. What is Klinefelter's syndrome?
16. Why would it be useful for an adopted child to have access to his or her birth parents' health histories?

15.3 Studying the Human Genome

HS-LS3-1, HS-ETS1-1

17. The first step in sequencing the human genome is
    a. locating overlapping sequences.
    b. identifying genes by finding promoters.
    c. cutting the DNA into manageable pieces.
    d. sorting introns from exons.
18. What is the role of restriction enzymes in studying the human genome?
    a. copying pieces of DNA
    b. labeling different nucleotides with chemical dyes
    c. separating different pieces of DNA based on their size
    d. cutting large DNA molecules into smaller pieces
19. The technique known as gel electrophoresis serves to
    a. bind DNA to chemical dyes.
    b. separate DNA fragments.
    c. reproduce DNA strands.
    d. synthesize nucleotide bases.
20. Approximately how many genes make up the human genome?
21. True or false: The functions of all genes in the human genome have been identified.
22. What benefits have resulted from the identification of SNPs associated with susceptibility to certain diseases or medical conditions?

23. In 2008, Congress passed the Genetic Information Nondiscrimination Act to prevent what?

24. What is the process of genomic imprinting?

CRITICAL THINKING

HS-LS3-1

25. **Plan an Investigation** Researchers suspect that a certain disease is caused by a recessive allele in a gene located on the X chromosome in fruit flies. Plan an investigation to test this hypothesis.

26. **Construct Tables** What are the possible genotypes of the parents of a child who is colorblind? Create a Punnett square to find out. Explain what the different possibilities are.

27. **Evaluate a Solution** Can a genetic counselor use a karyotype to identify a carrier of cystic fibrosis? Explain.

28. **Observe** The table shows the DNA sequences that are recognized by five different restriction enzymes and the locations where those enzymes cut. Which enzymes produce DNA fragments with "sticky ends"? What is the common feature of the sequences cut by these enzymes?

DNA Sequences Cut by Enzymes

Enzyme	Recognition Sequence
*Alu*I	A G ↓ C T T C ↑ G A
*Hae*III	G G ↓ C C C C ↑ G G
*Bam*HI	G ↓ G A T C C C C T A G ↑ G
*Hind*III	A ↓ A G C T T T T C G A ↑ A
*Eco*RI	G ↓ A A T T C C T T A A ↑ G

29. **Plan an Investigation** Fruit fly sex is determined by X and Y chromosomes, just as it is in humans. Researchers suspect that a certain disease is caused by a recessive allele in a gene located on the X chromosome in fruit flies. Design an experiment to test this hypothesis.

30. **Ask Questions** Review the table and compare the genome sizes and estimated number of genes for each organism or virus. What are some questions that you would ask to clarify the relationship between an organism or virus and its genome size? What questions would you need answered to determine why a single-celled organism has a larger genome size than other, more complex organisms?

Size Comparison of Various Genomes

Organism or Virus	Genome Size (bases)	Estimated Genes
Human (*Homo sapiens*)	3.2 billion	25,000
Laboratory mouse (*M. musculus*)	2.5 billion	24,174
Fruit fly (*D. melanogaster*)	165.0 million	13,600
Mustard weed (*A. thaliana*)	120.0 million	25,498
Roundworm (*C. elegans*)	97.0 million	19,000
Yeast (*S. cerevisiae*)	12.1 million	6294
Bacterium (*E. coli*)	4.6 million	4288
Human immunodeficiency virus (HIV)	9749.0	9

31. **Identify Variables** Identify what accounts for the vast differences of traits observed among different organisms, and for the complexity of these traits. Consider what you know about RNA transcription as part of your answer.

32. **Evaluate a Solution** Currently, tests are available for detecting genetic markers for different traits and syndromes. Evaluate the effectiveness of genetic testing as a solution for people facing the possibility of being a carrier for a genetic disorder. What are its limitations and advantages?

33. **Identify Patterns** Enzymes perform many different functions. Identify the different enzymes that have been mentioned in Chapters 13 through 15. What do they have in common? What are some of the different functions that they serve? What are some problems that an organism may face if its enzymes stopped working properly?

34. **Synthesize Information** How do human nondisjunction disorders explain how a male calico cat might be produced?

CROSSCUTTING CONCEPTS

35. **Connect to Technology** Private companies often announce advances in the technologies used for genetic testing. How have the technologies changed in the past twenty years? What new technologies are being developed currently? How do these differ from technologies used in the past?
36. **Cause and Effect** Explain why a small change to a gene can affect human traits. Give an example that illustrates why the change may bestow a benefit or may be harmful.
37. **Connect to Society** Perform research to find the concerns people have about genetic testing. What potentially positive and negative effects on society do you think the increasing availability of such testing will have?
38. **Cause and Effect** Restriction enzymes are named for the bacteria in which they were first discovered. For example, *Eco*RI was discovered in *E. coli*. Why do you think bacteria have DNA-snipping enzymes? (**Hint:** Think about the Hershey-Chase experiment.)

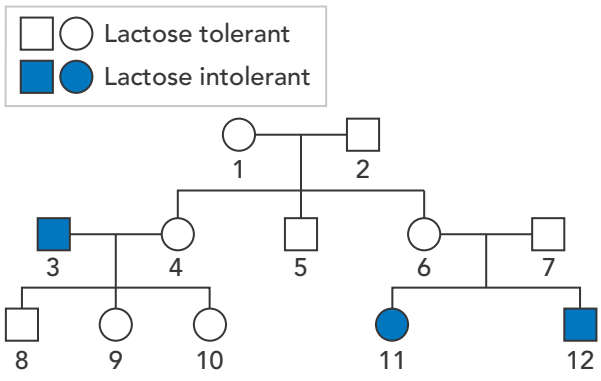
MATH CONNECTIONS

Analyze and Interpret Data

HS-LS3-1, CCSS.MATH.CONTENT.MP2

Refer to the pedigree chart to answer questions 39–41.

Lactose intolerance results from a person's inability to produce the enzyme lactase that breaks down the sugar lactose found in milk. Many adults experience lactose intolerance as they age and rely less on milk and dairy products as sources of nutrition. There is another rare form of lactose intolerance that affects infants from birth. A mutated version of the *LCT* gene that codes for the production of lactase exists as a recessive allele.



39. **Predict** Using your knowledge of Mendelian inheritance, create a Punnett square to show the possible genotypes of a child born to parents when the mother is homozygous dominant and the father is heterozygous for the *LCT* gene.
40. **Calculate** What is the ratio of the genotypes in the Punnett square you created? What is the ratio of the phenotypes?
41. **Reason Quantitatively** The pedigree shows the original parents numbered 1 and 2. They had three children. Compare the numbers in the ratios you determined to the appearance of the trait in the pedigree. Why were Persons 11 and 12 lactose intolerant while Persons 8, 9, and 10 were not?

LANGUAGE ARTS CONNECTION

Write About Science

HS-LS3-1, CCSS.ELA-LITERACY.WHST.9-10.5, CCSS.ELA-LITERACY.WHST.9-10.8

42. **Write Arguments** Should people have the right to keep genetic information private? Under what circumstances, if any, should scientists have access to genetic information without the person's consent? Present an argument to support your opinions.
43. **Plan and Revise** Explain the relationship between meiosis and Down syndrome, Turner's syndrome, and Klinefelter's syndrome. Make a plan, write a first draft, and revise your answer.

Read About Science

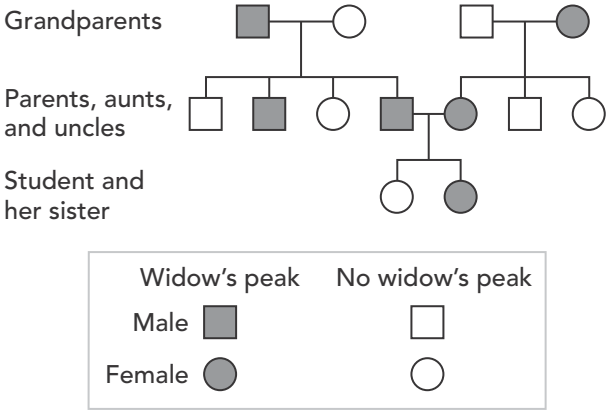
CCSS.ELA-LITERACY.RST.9-10.3, CCSS.ELA-LITERACY.RST.9-10.4

44. **Determine Meaning** Review the pedigree example in **Figure 15-7**. What is a pedigree? Determine the meaning of each symbol on the pedigree. What information do you need to already have that the pedigree does not provide in order to be able to understand it?
45. **Follow a Multistep Procedure** Review **Figures 15-12, 15-13,** and **15-14,** which show graphic representations of the steps of sequencing DNA. How would you describe the steps if you were to create a numbered list that someone else could follow to understand the procedure?

END-OF-COURSE TEST PRACTICE

For questions 1-2, refer to the following passage and diagram.

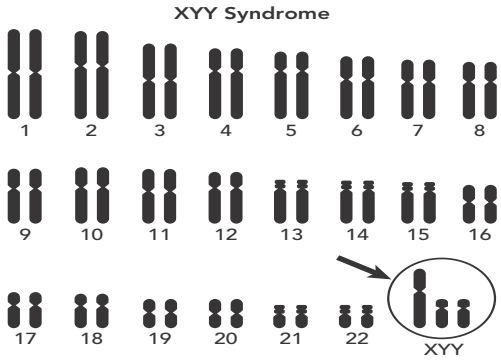
A student traced the recurrence of a widow's peak hairline in her family. Based on her interviews and observations, she drew the pedigree shown below.



1. Which pattern of inheritance is consistent with the pedigree?
   - **A**. sex-linked
   - **B**. multiple alleles
   - **C**. codominant alleles
   - **D**. recessive allele
   - **E**. dominant allele

2. What is the function of DNA in this pattern of inheritance?
   - **A**. Proteins containing DNA were passed from parent to offspring in each generation.
   - **B**. DNA was not folded properly in each of the affected individuals.
   - **C**. Mutations were induced in the widow's peak gene in each of the affected individuals.
   - **D**. A chromosome containing the allele for a widow's peak was passed from parent to offspring in each generation.
   - **E**. The chromosome containing the allele for a widow's peak was inactivated in each of the affected individuals.

3. The figure is a karyotype from a person with XYY Syndrome.



What event is MOST LIKELY to cause XYY Syndrome?
   - **A**. A duplication of the Y chromosome
   - **B**. Nondisjunction of chromosomes during meiosis
   - **C**. An error in DNA replication during mitosis
   - **D**. A change in a gene's DNA sequence
   - **E**. Inactivation of an X chromosome

4. How do alleles that display codominance differ from alleles that display simple dominance?
   - **A**. If two alleles are codominant then both alleles will be observed in heterozygotes.
   - **B**. If two alleles are codominant then the heterozygous phenotype will be somewhere between the homozygous phenotypes.
   - **C**. If two alleles display simple dominance then only the recessive allele will be observed.
   - **D**. If two alleles display simple dominance then neither allele will be observed in heterozygotes.
   - **E**. If two alleles display simple dominance then only the dominant allele is inheritable.

ASSESSMENT

For additional assessment practice, go online to access your digital course.

If You Had Trouble With…				
Question	1	2	3	4
See Lesson	15.1	15.1	15.2	15.1
Performance Expectation	HS-LS3-1	HS-LS3-1	HS-LS3-1	HS-LS3-1