

THEME FOCUS Patterns

The genetics of each organism is the basis of thwarsity within species.

(Idea Genetic technology improves human health and equality of life.

Section 1 • Applied Genetics

Section 2 • DNA Technology

Section 3 • The Human Genome

Section 1

Reading Preview

Essential Questions

- How is selective breeding used to produce organisms with desired traits?
- What are similarities and differences between inbreeding and hybridization?
- How does a Punnett square test cross help assess the genotypes of organisms?

Review Vocabulary

hybrid: organism that is heterozygous for a particular trait

New Vocabulary

selective breeding inbreeding test cross



Multilingual eGlossary

Figure 1 Dogs have traits that make them suited for different tasks: Saint Bernard—keen sense of smell; husky-endurance to run long distances; and German shepherd—high trainability.

Applied Genetics

Color Selective breeding is used to produce organisms with desired traits.

Real-World Reading Link Coin collectors separate rare coins from all other coins because the rare ones are more valuable. Just as certain coins are selected for their value, certain plants and animals have been selected and bred to produce organisms with traits that are valuable to humans.

Selective Breeding

You might be familiar with different breeds of dogs, such as Saint Bernards, huskies, and German shepherds. Observe some of the phenotypic traits of these breeds in Figure 1. All three have strong, muscular bodies. Saint Bernards have traits such as a keen sense of smell that make them good rescue dogs. Huskies are endurance runners and pull sleds long distances. German shepherds are highly trainable for special services.

Since ancient times, humans have bred animals with certain traits to obtain offspring that have desired traits. As a result, these traits become more common. Breeding for desired traits is not restricted to animals alone. Plants also are bred to produce desired traits, such as larger fruits and shorter growing times. The process by which desired traits of certain plants and animals are selected and passed on to their future generations is called selective breeding. Through the processes of hybridization and inbreeding, desired traits can be passed on to future generations.



Saint Bernard

Rescue dog



Husky Sled dog



German shepherd Service dog

Hybridization Recall that crossing parent organisms with different forms of a trait to produce offspring with specific traits results in hybrids. Farmers, animal breeders, scientists, and gardeners often use the production of hybrids, also known as hybridization. They select traits that will give hybrid organisms a competitive edge. These hybrid organisms can be bred to be more disease-resistant, to produce more offspring, or to grow faster. For example, plant breeders might choose to cross two different varieties of tomato plants in order to produce a hybrid that has both the disease resistance of one parent and the fast growth rate of the other parent.

Care must be taken to identify organisms with desired traits and successfully cross them to yield the right combination of traits from both parents. A disadvantage of hybridization is that it is time consuming and expensive. For example, it took rice breeders three decades to produce hybrid rice varieties that can produce higher yields than nonhybrid varieties. Because hvbrids can be bred to be more nutritious, to have the ability to adapt to a wide range of changes in the environment, and to produce greater numbers of offspring, the advantages of hybridization sometimes outweigh the disadvantages.

Inbreeding Once a breeder observes a desired trait in an organism, a process is needed to ensure that the trait is passed on to future generations. This process, in which two closely related organisms are bred to have the desired traits and to eliminate the undesired ones in future generations, is called inbreeding.

Pure breeds are maintained by inbreeding. Clydesdale horses, Angus cattle, and German shepherd dogs are all examples of organisms produced by inbreeding. You might have seen Clydesdale horses at parades and petting zoos. Horse breeders first bred the Clydesdale horse in Scotland hundreds of years ago for use as a farm horse. Because of their strong build, agility, and obedient nature, Clydesdales originally were inbred and used extensively for pulling heavy loads.

A disadvantage of inbreeding is that harmful recessive traits also can be passed on to future generations. Inbreeding increases the chance of homozygous recessive offspring. If both parents carry the recessive allele, the harmful trait likely will not be eliminated.



Reading Check Describe the disadvantages associated with hybridization and inbreeding.

JyJini Lab



Minil.ab

Model Hybridization

How are hybrid lilies produced? In this lab, you will examine techniques used by both professional plant breeders and amateur gardeners to produce the wide variety of lilies you might see growing in landscaped areas.

Procedure Z FI 3 E

- 1. Read and complete the lab safety form.
- 2. Obtain a labeled drawing of a lily flower and a fresh open lily flower. Examine the flower with a hand lens and identify the male anthers and the female pistil.
- 3. Use a cotton swab to gently rub an anther to pick up pollen.
- 4. Trade flowers with another lab group and, using the cotton swab, gently apply the pollen from your flower to the stigma of the pistil of the new flower.

Analysis

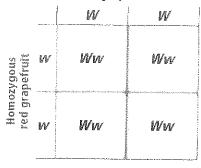
- 1. Infer When breeders hybridize lilies, they transfer pollen to the stigma of an unopened lily flower and then cover the stigma with a foil cap. Why do you think this would be necessary?
- 2. Think Critically A breeder produces a hybrid lily which then is allowed to grow and produce seeds naturally. When these seeds are planted, the new lily plants do not have the same characteristics as the hybrid parent. Hypothesize why this would occur.



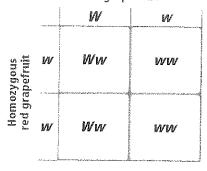
Launch Lab

Review Based on what you have read about selective breeding, how would you now answer the analysis questions?

Homozygous white grapefruit



Heterozygous white grapefruit



* Figure 2 The genotype of a white grapefruit tree can be determined by the results of a test cross with a homozygous red grapefruit.

Test Cross

An important thing a breeder has to determine when producing a hybrid is the genotype of the hybrid. Once a breeder observes the desired trait, if the trait is dominant, then the genotype of the organism could be homozygous dominant or heterozygous. The exact genotype is determined by performing a test cross. A test cross involves breeding an organism that has the unknown genotype with one that is homozygous recessive for the desired trait. If the parent's genotype is homozygous dominant, all the offspring will have the dominant phenotype; if it is heterozygous, the offspring will show a 1:1 phenotypic ratio.

Performing a test cross Suppose a breeder wants to produce hybrid white grapefruits. In grapefruit trees, white fruit color is the dominant trait, while red is recessive. Therefore, the red grapefruit trees in the orchard must be homozygous recessive (ww). The genotype of the hybrid white grapefruit tree obtained by the breeder can be homozygous dominant (WW) or heterozygous (Ww) for the white color. Therefore, the breeder has to perform a test cross to determine the genotype of the white grapefruit tree. Remember when performing a cross, pollen from the flower of one plant is transferred to the female organ in a flower of another plant.

Results As shown in the top Punnett square in **Figure 2**, if the white grapefruit tree is homozygous dominant (*WW*) and is crossed with a red grapefruit tree (*ww*), then all the offspring will be heterozygous (*Ww*) and white in color. In this case, all of the offspring will have the dominant phenotype. However, as shown in the second Punnett square in **Figure 2**, if the white grapefruit tree is heterozygous (*Ww*), then half the number of offspring will be white and half will be red, and the phenotypic ratio will be 1:1. Review the results in the Punnett squares in **Figure 2**. If the white grapefruit tree is homozygous, all offspring will be heterozygous—white in color. If the tree is heterozygous, half of the test-cross offspring will be white and half will be red.

Section 1 Assessment

Section Summary

- Selective breeding is used to produce organisms with traits that are considered desirable.
- Hybridization produces organisms with desired traits from parent organisms with different traits.
- Inbreeding creates pure breeds.
- A test cross can be used to determine an organism's genotype.

Understand Main Ideas

- **1.** Assess the effect of selective breeding on food crops.
- **2. Describe** three traits that might be desired in sheep. How can these traits be passed on to the next generation? Explain.
- 3. Compare and contrast inbreeding and hybridization.
- **4. Predict** the phenotype of offspring from a test cross between a seedless orange (ss) and an orange with seeds (Ss).

Think Critically

5. Evaluate Should a cow and a bull that both carry recessive alleles for a mutation that causes decreased milk production be bred? Why or why not?

MATHITY Biology

6. A breeder performs a test cross to determine the genotype of a black cat. He crosses the black cat (*BB* or *Bb*) with a white cat (*bb*). If 50 percent of the offspring are black, what is the genotype of the black cat?



Section 2

Reading Preview

Essential Guestions

- What are the different tools and processes used in genetic engineering?
- How does genetic engineering manipulate recombinant DNA?
- What are the similarities between selective breeding and genetic engineering?
- How can genetic engineering and biotechnology be used to improve human life?

Review Vocabulary

DNA: the genetic material of all organisms, composed of two complementary chains of nucleotides wound in a double helix

New Vocabulary

genetic engineering
genome
restriction enzyme
gel electrophoresis
recombinant DNA
plasmid
DNA ligase
transformation
cloning
polymerase chain reaction
transgenic organism



Figure 3 The gene for green fluorescent protein (GFP) was introduced into mosquito larvae so that researchers could verify that exogenous DNA was inserted.

Predict how genetic engineering might be used in the future by the medical field.

DNA Technology

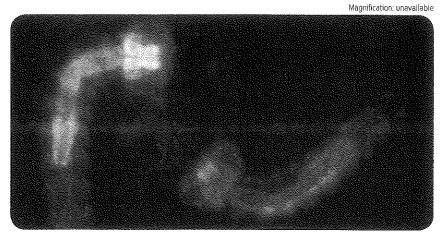
Researchers use genetic engineering to manipulate DNA.

Real-World Reading Link Have you seen a handmade patchwork quilt? Patchwork quilts are created by combining different pieces of fabric. Scientists use a similar process and combine DNA from different sources to create an organism with unique traits.

Genetic Engineering

By about 1970, researchers had discovered the structure of DNA and had determined the central dogma that information flowed from DNA to RNA and from RNA to proteins. However, scientists did not know much about the function of individual genes. Suppose your friend told you the final score of a high school football game but did not tell you how each player contributed to the game. Your curiosity about the details of the game is similar to the curiosity scientists experienced because they did not know how each gene contributed to a cell's function.

The situation changed when scientists began using **genetic engineering**, technology that involves manipulating the DNA of one organism in order to insert exogenous DNA (the DNA of another organism). For example, researchers have inserted a gene for a bioluminescent protein called green fluorescent protein (GFP) into various organisms. GFP, which is a substance naturally found in jellyfishes that live in the north Pacific Ocean, emits a green light when it is exposed to ultraviolet light. Organisms that have been genetically engineered to synthesize the DNA for GFP, such as the mosquito larvae shown in **Figure 3**, can be easily identified in the presence of ultraviolet light. The GFP DNA is attached to exogenous DNA to verify that the DNA has been inserted into the organism. These genetically engineered organisms are used in various processes, such as studying the expression of a particular gene, investigating cellular processes, studying the development of a certain disease, and selecting traits that might be beneficial to humans.



Genetically engineered mosquito larvae

VOCABULARY

Broggiano, via das a ve

Manipulate

to manage or utilize skillfully Scientists use technology to manipulate genetic information in order to test scientific hypotheses.

DNA Tools

You have learned that selective breeding is used to produce plants and animals with desired traits. Genetic engineering can be used to increase or decrease the expression of specific genes in selected organisms. It has many applications from human health to agriculture.

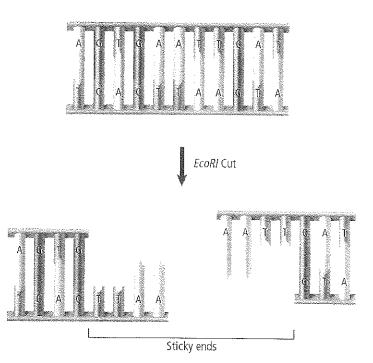
An organism's **genome** is the total DNA present in the nucleus of each cell. As you will learn in the next section, genomes, such as the human genome, can contain millions and millions of nucleotides. In order to study a specific gene, DNA tools can be used to manipulate DNA and to isolate genes from the rest of the genome.

Restriction enzymes Some types of bacteria contain powerful defenses against viruses. These cells contain proteins called **restriction enzymes** that recognize and bind to specific DNA sequences and cleave the DNA within that sequence. A restriction enzyme, also called an endonuclease (en doh NEW klee ayz), cuts the viral DNA into fragments after it enters the bacteria. Since their discovery in the late 1960s, scientists have identified and isolated hundreds of restriction enzymes. Restriction enzymes are used as powerful tools for isolating specific genes or regions of the genome. When the restriction enzyme cleaves genomic DNA, it creates fragments of different sizes that are unique to every individual.

EcoRi One restriction enzyme that is used widely by scientists is known as *EcoRI*. As illustrated in **Figure 4**, *EcoRI* specifically cuts DNA containing the sequence GAATTC. The ends of the DNA fragments created by *EcoRI* are called sticky ends because they contain single-stranded DNA that is complementary. The ability of some restriction enzymes to create fragments with sticky ends is important because these sticky ends can be joined together with other DNA fragments that have complementary sticky ends.



Reading Check Generalize how restriction enzymes are used.



* **Figure 4** DNA containing the sequence GAATTC can be cut by the restriction enzyme *EcoR*i to produce sticky ends.

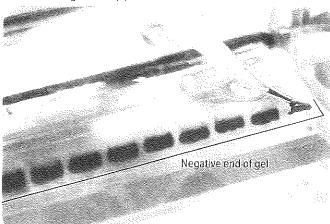


Animation



(I)Klaus Guldbrandsen/Photo Researchers; (r)NOAA

Loading the gel Solution containing DNA is dropped into holes at one end of the gel with a pipette.



Fragment pattern A staining solution binds to the separated DNA fragments in the gel, making them visible under ultraviolet light.

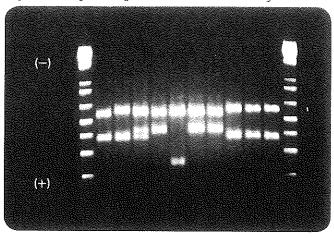


Figure 5 When the loaded gel is placed in an electrophoresis tank and the electric current is turned on, the DNA fragments separate.

However, not all restriction enzymes create sticky ends. Some enzymes produce fragments containing blunt ends–created when the restriction enzyme cuts straight across both strands. Blunt ends do not have regions of single-stranded DNA and can join to any other DNA fragment with blunt ends.

Gonnection ত আনুষ্টেভ Gel electrophoresis An electric current is used to separate DNA fragments according to the size of the fragments in a process called gel electrophoresis. Figure 5 shows how the DNA fragments are loaded on the negatively charged end of a gel. When an electric current is applied, the DNA fragments move toward the positive end of the gel. The smaller fragments move farther faster than the larger ones. The unique pattern created based on the size of the DNA fragment can be compared to known DNA fragments for identification. Also, portions of the gel containing each band can be removed for further study.

JyJiniLab

Model Restriction Enzymes



MiniLab

How are sticky ends modeled? Use scissors and tape to produce paper DNA fragments with sticky ends and a recombinant DNA plasmid.

Procedure Zz



- 1. Read and complete the lab safety form.
- 2. Obtain one straight paper DNA sequence from your teacher, which will represent genomic DNA, and one circular paper DNA sequence, which will represent a plasmid.
- 3. Find each GAATTC sequence recognized by the restriction enzyme EcoRI and cleave the genome and plasmid DNA using scissors.
- 4. Use tape to make a recombinant DNA plasmid.

Analysis

- 1. Compare your plasmid to those made by other lab groups. How many different recombinant plasmids could be made using this particular genomic sequence? Explain.
- 2. Infer what enzyme was represented by the scissors. Explain.

Recombinant DNA Technology

When DNA fragments have been separated by gel electrophoresis, fragments of a specific size can be removed from the gel and combined with DNA fragments from another source. This newly generated DNA molecule, with DNA from different sources, is called **recombinant DNA**. Recombinant DNA technology has revolutionized the way scientists study DNA because it enables individual genes to be studied.

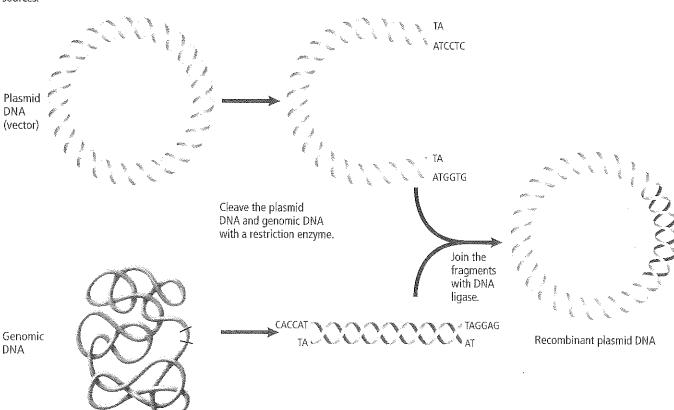
Large quantities of recombinant DNA molecules are needed in order to study them. A carrier, called a vector, transfers the recombinant DNA into a bacterial cell called the host cell. Plasmids and viruses are commonly used vectors. **Plasmids**—small, circular, double-stranded DNA molecules that occur naturally in bacteria and yeast cells—can be used as vectors because they can be cut with restriction enzymes. If a plasmid and a DNA fragment obtained from another genome have been cleaved by the same restriction enzyme, the ends of each DNA fragment will be complementary and can be combined, as shown in **Figure 6.** An enzyme normally used by cells in DNA repair and replication, called **DNA ligase**, joins the two DNA fragments chemically. Ligase joins DNA fragments that have sticky ends as well as those that have blunt ends.

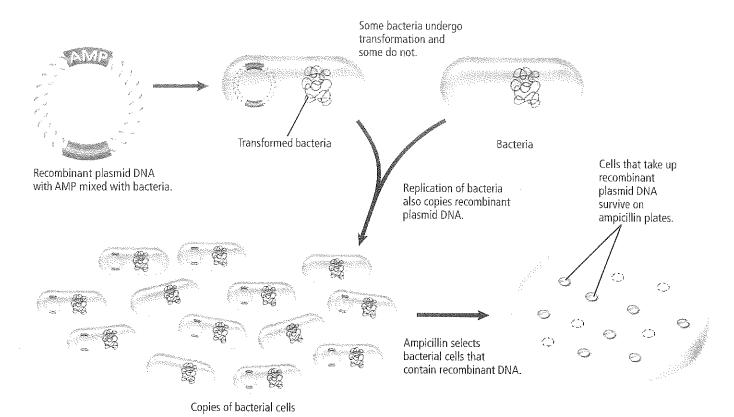
Examine **Figure 6** again. Notice that the resulting circular DNA molecule contains the plasmid DNA and the DNA fragment isolated from another genome. This recombinant plasmid DNA molecule now can be inserted into a host cell so that large quantities of this type of recombinant DNA can be made.



Reading Check Relate restriction enzymes to recombinant DNA.

Figure 6 Recombinant DNA is created by joining together DNA from two different sources.





Gene cloning To make a large quantity of recombinant plasmid DNA, bacterial cells are mixed with recombinant plasmid DNA. Some of the bacterial cells take up the recombinant plasmid DNA through a process called transformation, as shown in Figure 7. Bacterial cells can be transformed using electric pulsation or heat. Recall that all cells, including bacterial cells, have plasma membranes. A short electric pulse or a brief rise in temperature temporarily creates openings in the plasma membrane of the bacteria. These temporary openings allow small molecules, such as the recombinant plasmid DNA, to enter the bacterial cell. The bacterial cells make copies of the recombinant plasmid DNA during cell replication. Large numbers of identical bacteria, each containing the inserted DNA molecules, can be produced through this process called cloning.

Recombinant plasmid DNA contains a gene that codes for resistance to an antibiotic such as ampicillin (AMP). Researchers use this gene to distinguish between bacterial cells that have taken up the recombinant plasmid DNA and those that have not. Notice in Figure 7 that when the transformed bacterial cells are exposed to the specific antibiotic, only the bacterial cells that have the plasmid survive.

DNA sequencing The sequence of the DNA nucleotides of most organisms is unknown. Knowing the sequence of an organism's DNA or of a cloned DNA fragment provides scientists with valuable information for further study. The sequence of a gene can be used to predict the function of the gene, to compare genes with similar sequences from other organisms, and to identify mutations or errors in the DNA sequence. Because the genomes of most organisms are made up of millions of nucleotides, the DNA molecules used for sequencing reactions first must be cut into smaller fragments using restriction enzymes.

Figure 7 Clones containing copies of the recombinant DNA can be identified and used for further study when the bacterial cells that do not contain recombinant DNA die.

VOCABULARY

SCHERCE USAGE U. COMMON USAGE

Transformation

Science usage: process by which one type of bacterium takes up the DNA from another source Transformation of bacteria involves the uptake of plasmid DNA.

Common usage: the act of change The transformation of the room was complete with the addition of new drapes.

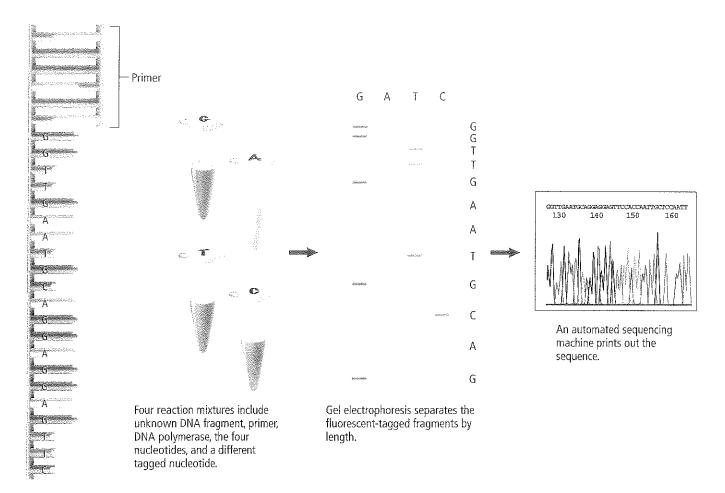


 Figure 8 DNA can be seguenced using fluorescent-tagged nucleotides. Describe how the sequence of the

original DNA template is determined.

Follow **Figure 8** to understand how DNA is sequenced. Scientists mix an unknown DNA fragment, DNA polymerase, and the four nucleotides—A, C, G, T—in a tube. A small amount of each nucleotide is tagged with a different color of fluorescent dye, which also modifies the structure of the nucleotide. Every time a modified fluorescenttagged nucleotide is incorporated into the newly synthesized strand, the reaction stops. This produces DNA strands of different lengths. The sequencing reaction is complete when the tagged DNA fragments are separated by gel electrophoresis. The gel is then analyzed in an automated DNA sequencing machine that detects the color of each tagged nucleotide. The sequence of the original DNA template is determined from the order of the tagged fragments.

Polymerase chain reaction Once the sequence of a DNA fragment is known, a technique called the **polymerase chain reaction** (PCR) can be used to make millions of copies of a specific region of a DNA fragment. PCR is extremely sensitive and can detect a single DNA molecule in a sample. PCR is useful because this single DNA molecule then can be copied, or amplified, numerous times to be used for DNA analysis. Follow **Figure 9** as you read about the steps of PCR.

Step 1 PCR is performed by placing the DNA fragment to be copied, DNA polymerase, the four DNA nucleotides, and two short singlestranded pieces of DNA called primers in a tube. The primers are complementary to the ends of the DNA fragment that will be copied and used as starting points for DNA synthesis. PCR begins when the tube is heated.

Step 2 The heat separates the two strands of the template DNA fragment. When the tube is cooled, the primers can bind to each strand of the template DNA. An automated machine called a thermocycler is used to cycle the tube containing all of the components involved in PCR through various hot and cool temperatures.

Step 3 As shown in Figure 9, each primer is made to bind to one strand of the DNA fragment. Once the primers are bound, DNA polymerase incorporates the correct nucleotides between the two primers as in DNA replication. This process of heating, cooling, and nucleotide incorporation is repeated 20 to 40 times, resulting in millions of copies of the original fragment. Because the separation of DNA strands requires heat, the DNA polymerase used in PCR has to be able to withstand high temperatures. This special DNA polymerase was isolated from a thermophilic, or heat-loving, bacterium such as those found living in the hot springs of Yellowstone National Park.

Because PCR can detect a single DNA molecule in a sample, it has become one of the most powerful tools used by scientists. PCR is not used only by researchers in laboratories, but also by forensic scientists to identify suspects and victims in crime investigations, and by doctors to detect infectious diseases, such as AIDS.

Figure 9 PCR is a biological version of a copy machine. During each PCR cycle, the reaction mixture is heated to separate the DNA strands and then cooled to allow primers to bind to complementary sequences. The DNA polymerase then adds nucleotides to form new DNA molecules.



Animation

Reading Check Describe the polymerase chain reaction using an analogy.

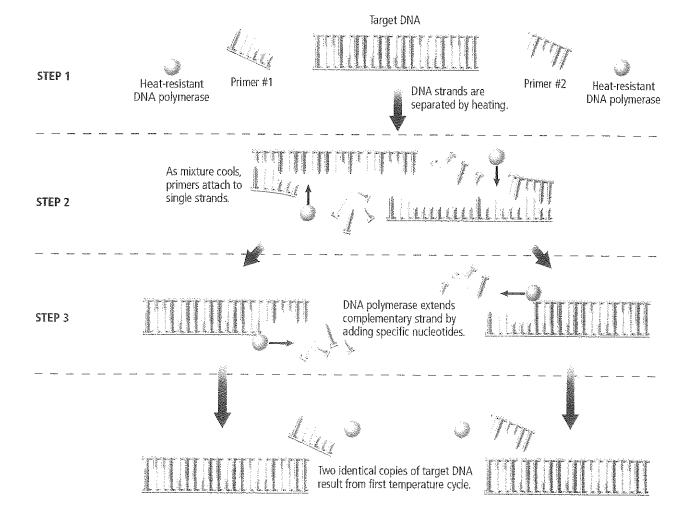


Table 1	Genetic Engineering	Interactive Table
Tool/Process	Surgion	Applications
Restriction enzymes Ex: EcoRl	Cut DNA strands into fragments	Used to create DNA fragments with sticky ends or blunt ends that can join with other DNA fragments
Gel electrophoresis	Separates DNA fragments by size	Used to study DNA fragments of various sizes
Recombinant DNA tech- nology	Combines a DNA fragment with DNA from another source (exogenous DNA)	Used to create recombinant DNA to be used to study individual genes and genetically engineered organisms, and in the treatment of certain diseases
Gene cloning	Produces large numbers of identical recombinant DNA molecules	Used to create large amounts of recombinant DNA to be used in genetically engineered organisms
DNA sequencing	Identifies the DNA sequence of cloned recombinant DNA molecules for further study	Used to identify errors in the DNA sequence, to predict the function of a particular gene, and to compare to other genes with similar sequences from different organisms
Polymerase chain reaction (PCR)	Makes copies of specific regions of sequenced DNA	Used to copy DNA for any scientific investigation, including forensic analysis and medical testing

Genetic engineering uses powerful tools, summarized in **Table 1**, to study and manipulate DNA. Although researchers investigate many different problems, their experimental procedures often include cleavage by a restriction enzyme, isolation of fragments, combination with exogenous DNA, cloning or PCR, and identification of sequences.

Biotechnology

Biotechnology—the use of genetic engineering to find solutions to problems—makes it possible to produce organisms that contain individual genes from another organism. Recall that organisms such as the mosquito larvae shown in **Figure 3** have a gene from another organism. Such organisms, genetically engineered by inserting a gene from another organism, are called **transgenic organisms**. Transgenic animals, plants, and bacteria are used not only for research, but also for medical and agricultural purposes.

Transgenic animals Currently, scientists produce most transgenic animals in laboratories for biological research. Mice, fruit flies, and the roundworm *Caenorhabditis elegans*, also called *C. elegans*, are widely used in research laboratories around the world to study diseases and develop ways to treat them. Some transgenic organisms, such as transgenic livestock, have been produced to improve the food supply and human health. Transgenic goats have been engineered to secrete a protein called antithrombin III, which is used to prevent human blood from forming clots during surgery. Researchers are working to produce transgenic chickens and turkeys that are resistant to diseases. Several species of fishes also have been genetically engineered to grow faster. In the future, transgenic organisms might be used as a source of organs for organ transplants.

CARRESS IN BIOLOGY

Geneticist Using many of the DNA tools, a geneticist might research genes, inheritance, and the variations of organisms. Some geneticists are medical doctors who diagnose and treat genetic conditions.

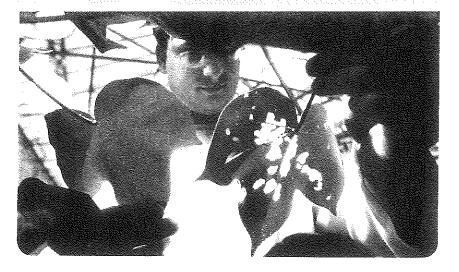


Figure 10 This researcher is examining cotton plant leaves. The leaf on the left has been genetically engineered to resist insect infestation.

Transgenic plants Many species of plants have been genetically engineered to be more resistant to insect or viral pests. In 2006, about 69.9 million hectares grown by 7 million farmers in 18 countries were planted with transgenic crops. These crops included herbicide- and insecticide-resistant soybeans, corn, cotton, and canola. Scientists now are producing genetically engineered cotton, as shown in Figure 10, that resists insect infestation of the bolls. Researchers also are developing peanuts and soybeans that do not cause allergic reactions.

Other crops are being grown commercially and being fieldtested. These crops include sweet-potato plants that are resistant to a virus that could kill most of the African harvest, rice plants with increased iron and vitamins that could decrease malnutrition in Asian countries, and a variety of plants able to survive extreme weather conditions. Prospective crops include bananas that produce vaccines for infectious diseases, such as hepatitis B, and plants that produce biodegradable plastics.

Transgenic bacteria Insulin, growth hormones, and substances that dissolve blood clots are made by transgenic bacteria. Transgenic bacteria also slow the formation of ice crystals on crops to protect them from frost damage, clean up oil spills more efficiently, and decompose garbage.

Section 2 Assessment

Section Summary

- Genetic engineering is used to produce organisms that are useful to humans.
- Recombinant DNA technology is used to study individual genes.
- DNA fragments can be separated using gel electrophoresis.
- Clones can be produced by transforming bacteria with recombinant DNA.
- The polymerase chain reaction is used to make copies of small DNA sequences.
- Transgenic organisms are being created to increase the quality of human life.

Understand Main Ideas

- 1. Sequence how recombinant DNA is made and manipulated.
- 2. Explain why some plasmids contain a gene for resistance to an antibiotic.
- 3. Describe how genetic engineering can improve human health.
- 4. Contrast one major difference between selective breeding and genetic engineering.

Think Critically

5. Evaluate Several popular movies and books involve mutated organisms. Are these transgenic organisms a possibility? Why or why not?

Wassingto Biology

6. Why would a business synthesize and sell DNA? Who would their customers be? Write a list of possible uses for DNA that is synthesized in a laboratory.



Section 3

Reading Preview

Essential Questions

- What are the components of the human genome?
- How do forensic scientists use DNA fingerprinting?
- How can information from the human genome be used to treat human diseases?

Review Vocabulary

codon: the triplet of bases in the DNA or mRNA

New Vocabulary

DNA fingerprinting bioinformatics DNA microarray single nucleotide polymorphism haplotype pharmacogenomics gene therapy genomics proteomics



The Human Genome

Genomes contain all of the information needed for an organism to grow and survive.

Real-World Reading Link When you put together a jigsaw puzzle, you might first find all the border pieces and then fill in the other pieces. Sequencing the human genome can be compared to putting together a jigsaw puzzle. Just as you have to figure out which puzzle pieces fit together, scientists had to determine the sequence of the base pairs along the length of a human chromosome.

The Human Genome Project

The Human Genome Project (HGP) was an international project that was completed in 2003. A genome is the complete genetic information in a cell. The goal of the HGP was to determine the sequence of the approximately three billion nucleotides that make up human DNA and to identify all of the human genes. If all the nucleotides in the human genome were the size of the type on this page and fused together in one continuous line, the line would extend from Los Angeles, California, to Panama, as illustrated in **Figure 11.**

Though the HGP is finished, analysis of the data generated from this project will continue for many decades. To complete this huge task, researchers also have studied the genomes of several other organisms, including the fruit fly, the mouse, and *Escherichia coli*—the bacterium present in the human intestines. Studies in nonhuman organisms help to develop the technology required to handle the large amounts of data produced by the Human Genome Project. These technologies help to interpret the function of newly identified human genes.

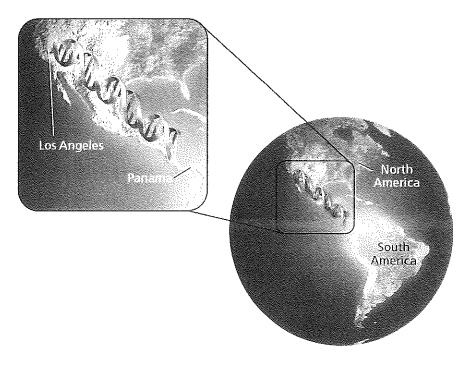


Figure 11 If all the DNA in the human genome were fused together in one continuous line, it would stretch from California to Panama.



Decodingthehumansurntodgenomeseque ncecanhfgeteirunfhdbecomparedtorefdt wiqppnbfreadingabookthatwaswregdfst wuthnbkutiprintedlhjgkkkkincorrectlyima ginethegenomeasterdlongpmllwordstkfh gnviinabooknvhgytpwmlwrittenwithoutc apitalizationkghtowkfgcbvjorpunctuation hgitofcjwithoutvhtofutibreakshkovpabet weenwordssentencesorvhgotwpqmnkpar agraphsandwithfoagwitostringsofletters dhfiruwqscatteredbetweenandwithin sentencesinghomlaordertohdqpvunderstandwhatiswrittenthejumbledbghfqomkslte xthastobeghqpmsddecoded.

* Figure 12 The genetic information contained within the human genome has to be decoded in order to uncover important sequences.

interpret the text by decoding the jumbled sentences.

Sequencing the genome Human DNA is organized into 46 chromosomes. In order to determine one continuous human genome sequence, each of the 46 human chromosomes was cleaved. Several different restriction enzymes were used in order to produce fragments with overlapping sequences. These fragments were combined with vectors to create recombinant DNA, cloned to make many copies, and sequenced using automated sequencing machines. Computers analyzed the overlapping regions to generate one continuous sequence.

Decoding the sequence of the human genome can be compared to reading a book that was printed in code. Imagine the genome as words in a book written without capitalization, punctuation, or breaks between words, sentences, or paragraphs. Suppose there are strings of letters scattered between and within sentences. **Figure 12** illustrates how a page from such a book might look. In order to understand what is written, you have to decode the jumbled text. Similarly, scientists had to decode the genetic code in the human genome.

After sequencing the entire human genome, scientists observed that less than two percent of all of the nucleotides in the human genome code for all the proteins in the body. That is, the genome is filled with long stretches of repeated sequences that have no direct function. These regions are called noncoding sequences.

DNA fingerprinting Unlike the protein-coding regions of DNA that are almost identical among individuals, the long stretches of noncoding regions of DNA are unique to each individual. When these regions are cut by restriction enzymes, as described earlier in this chapter, the set of DNA fragments produced is unique to every individual. **DNA fingerprinting** involves separating these DNA fragments using gel electrophoresis in order to observe the distinct banding patterns that are unique to every individual. Forensic scientists use DNA fingerprinting to identify suspects and victims in criminal cases, to determine paternity, and to identify soldiers killed in war.

VOCABULARY

有打造自新物质的 计特别数据 医多子

Sequence (SEE kwens)

a continuous series

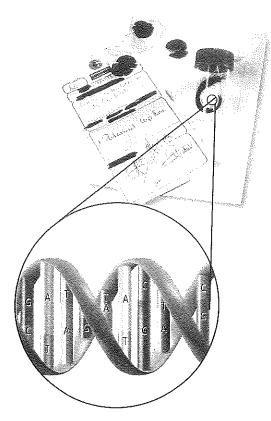
The sequence of colors formed a
beautiful pattern.



Virtual Lab

CAREERS IN BROKENS

Forensic Scientist Genetic engineering is a technology used widely by forensic scientists. They use the various tools and processes, such as DNA fingerprinting, in criminal and archaeological investigations.



* **Figure 13** People can be identified using the genetic information contained in blood, hair, semen, or skin.

Figure 13 shows a sample obtained from hair that forensic scientists can use for DNA fingerprinting. PCR is used to copy this small amount of DNA to create a larger sample for analysis. The amplified DNA then is cut using different combinations of restriction enzymes. The fragments are separated by gel electrophoresis and compared to DNA fragments from known sources, such as victims and suspects in a criminal case, to locate similar fragmentation patterns. There is a high probability that the two DNA samples came from the same person if two fragmentation patterns match. Since its development in England in 1985, DNA fingerprinting has been used not only to convict criminals but also to free innocent people who had been wrongfully imprisoned. Figure 14 provides a closer look at the history of genetic technology.



Reading Check **Summarize** how forensic scientists use DNA fingerprinting.

Identifying Genes

Once the genome has been sequenced, the next step in the process is to identify the genes and determine their functions. The functions of many of the genes in the human genome are still unknown. Researchers use techniques that integrate computer analysis and recombinant DNA technology to determine the function of these genes.

For organisms such as bacteria and yeast, whose genomes do not have large regions of noncoding DNA, researchers have identified genes by scanning the sequence for open reading frames (or ORFs, pronounced "orphs"). ORFs are stretches of DNA containing at least 100 codons that begin with a start codon and end with a stop codon. While these sequences might indicate a gene, they will be tested to determine if these sequences produce functioning proteins.

Figure 14Discoveries in Genetics

Many studies in genetics have led to advances in biotechnology.



1983 Kary Mullis invents the polymerase chain reaction, for which he will be awarded the Nobel Prize in 1993.

(2)51)

10078

de ar

1959 Down syndrome is the first chromosomal abnormality identified in humans. 1972 Paul Berg creates the first recombinant DNA molecules.

1973 Herbert Boyer, Annie Chang, Stanley Cohen, and Robert Helling discover that recombinant DNA reproduce if inserted into bacteria. (t)Pascal Goetgheluck/Photo Researchers; (b)Pies

Recall that a codon is a group of three nucleotides that code for an amino acid. Researchers look for the start codon AUG and a stop codon such as UAA, UGA, or UAG. ORF analysis has been used to identify correctly over 90 percent of genes in yeast and bacteria. However, the identification of genes in more complex organisms such as humans requires more sophisticated computer programs called algorithms. These algorithms use information, such as the sequence of the genomes of other organisms, to identify human genes.

Bioinformatics

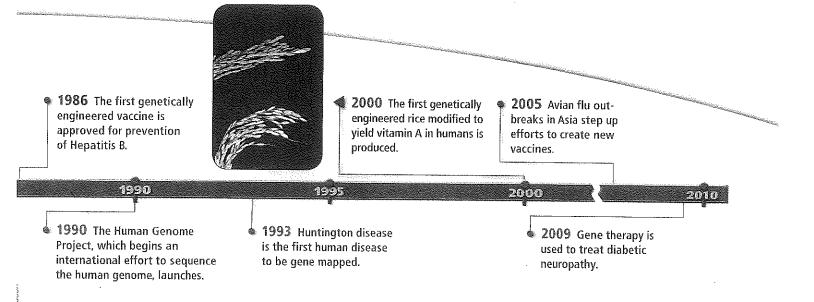
The completion of the HGP and the sequencing of the genomes of other organisms have resulted in large amounts of data. Not only has this enormous amount of data required careful storage, organization, and indexing of sequence information, but it also has created a new field of study. This field of study, called **bioinformatics**, involves creating and maintaining databases of biological information. The analysis of sequence information involves finding genes in DNA sequences of various organisms and developing methods to predict the structure and function of newly discovered proteins. Scientists also study the evolution of genes by grouping protein sequences into families of related sequences and comparing similar proteins from different organisms.

DNA Microarrays

Analyzing all the expressed genes from a given organism or a specific cell type can be useful. This analysis can be done using **DNA microarrays**, which are tiny microscope slides or silicon chips that are spotted with DNA fragments. DNA microarrays can contain a few genes, such as the genes that control the cell cycle, or all of the genes of the human genome. Therefore, a large amount of information can be stored in one small slide or chip. DNA microarrays help researchers determine whether the expression of certain genes is caused by genetic factors or environmental factors.

Study Tip

BioJournal As you read about the human genome, list several beneficial uses of this information.



Follow the steps involved in doing the DNA microarray experiment shown in **Figure 15.** mRNA from two different populations of cells is isolated and converted into complementary DNA (cDNA) strands using an enzyme called reverse transcriptase. The complementary DNA from each cell population is labeled with a specific fluorescent dye—for example, red for cancer cells and green for normal cells. Both pools of complementary DNA are combined on the microarray slide and incubated.

Figure 15 shows the fluorescent signals that are produced when the microarray slide is analyzed. When the expression of a gene is the same in both the normal and cancer cells, a yellow spot is produced on the chip. If the expression of a gene is higher in cancer cells, then the spot formed is red. However, if the expression is higher in normal cells, then the spot formed is green.

Because one DNA microarray slide can contain thousands of genes, researchers can examine changes in the expression patterns of multiple genes at the same time. Scientists also are using DNA microarrays to identify new genes and to study changes in the expression of proteins under different growth conditions.

The Genome and Genetic Disorders

Although more than 99 percent of all nucleotide base sequences are exactly the same in all people, sometimes there are variations that are linked to human diseases. These variations in the DNA sequence that occur when a single nucleotide in the genome is altered are called **single nucleotide polymorphisms** or SNPs. For a variation to be considered an SNP, it must occur in at least one percent of the population. Many SNPs have no effect on cell function, but scientists hypothesize that SNP maps will help identify many genes associated with many different types of genetic disorders.

DATA ANALYSIS LAB



Apply Concepts

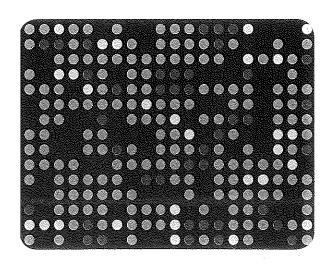
How can DNA microarrays be used to classify types of prostate cancer? The gene expression profiles between normal prostate cells and prostate cancer cells can be compared using DNA microarray technology.

Data and Observations

The diagram shows a subset of the data obtained.

Think Critically

- **1. Calculate** the percentage of spots that are yellow. Then calculate the percentage of green spots and red spots.
- 2. Explain why some of the spots are black.
- **3. Apply Concepts** How would you choose a gene to study as a cause of prostate cancer?



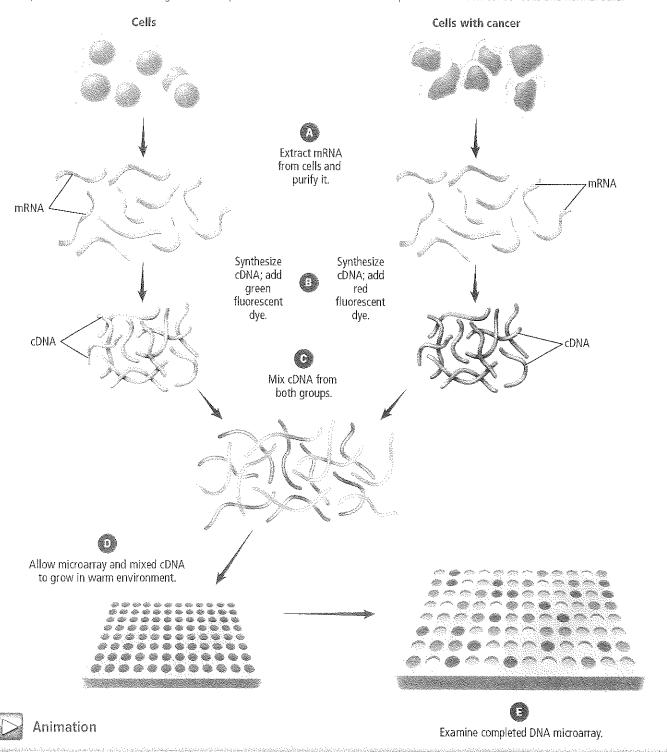
*Data obtained from: Lapointe, et al. 2004. Gene expression profiling identifies clinically relevant subtypes of prostate cancer. PNAS 101: 811–816.



Visualizing Microarray Analysis

Figure 15

In this experiment, the expression of thousands of human genes was detected by DNA microarray analysis. Each spot on the microarray chip represents a gene. A red spot indicates the expression of a gene is higher in cancer cells compared to normal cells. A green spot indicates the expression in normal cells is higher. Yellow spots indicate no difference in the expression between cancer cells and normal cells.



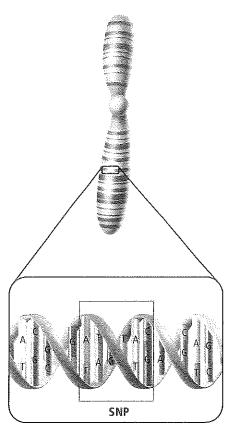
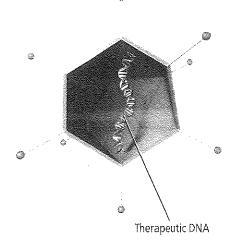


Figure 16 The HapMap project involves grouping all adjacent SNPs that are inherited together into haplotypes.

** Figure 17 DNA can be encapsulated in a virus and delivered into a patient to replace a defective gene. Once the virus enters the cells, the genetic information is released into the nucleus and inserted into the genome.



The HapMap project An internationl group of scientists is creating a catalog of common genetic variations that occur in humans. Linked genes are inherited together and similarly, genetic variations located close together also tend to be inherited together. Therefore, regions of linked variations in the human genome, known as haplotypes, can be located. The project to create this catalog is called the haplotype map, or HapMap project. Assembling the HapMap involves identifying groups of SNPs in a specific region of DNA.

Figure 16 shows how the genome is divided into haplotypes. Once completed, the HapMap will describe what these variations are, where they occur in our DNA, and how they are distributed among people within populations and among populations in different parts of the world. This information will help researchers find genes that cause disease and affect an individual's response to drugs.

Pharmacogenomics Sequencing the human genome combines the knowledge of genes, proteins, and SNPs with other areas of science. The study of how genetic inheritance affects the body's response to drugs is called **pharmacogenomics** (far muh koh jeh NAW mihks). The benefits of pharmacogenomics include more accurate dosing of drugs that are safer and more specific. Researchers hope that pharmacogenomics will allow for drugs to be custom-made for individuals based on their genetic makeups. Prescribing drugs based on an individual's genetic makeup will increase safety, speed recovery, and reduce side effects. Perhaps one day when you are sick, your doctor will read your genetic code and prescribe medicine tailor-made for you.

Gene therapy A technique aimed at correcting mutated genes that cause human diseases is called **gene therapy.** Scientists insert a normal gene into a chromosome to replace a dysfunctional gene. In most gene therapy studies, inserting a normal gene into a viral vector, like the one in **Figure 17**, produces recombinant DNA. Target cells in the patient are infected with the virus and the recombinant DNA material is released into the affected cells. Once deposited into cells, the normal gene inserts itself into the genome and begins functioning.

Connection to In 1990, the first clinical gene therapy trial at the National Institutes of Health was conducted on a four year old child with severe combined immunodeficiency (SCID). The Food and Drug Administration (FDA) monitors new medical trials, including gene therapy. Gene therapy has seen its share of setbacks, but the possibilities are endless when it comes to new treatments. Recent gene therapy trials include work with diabetes, cancer, retinal disease, Parkinson's disease, and others.



Reading Check **Compare and contrast** pharmacogenomics to gene therapy.

Genomics and Proteomics

Sequencing the human genome began what researchers call "the genomic era." **Genomics** is the study of an organism's genome. Genomics has become one of the most powerful strategies for identifying human genes and interpreting their functions. In addition to the mass of data obtained from sequencing the genomes of multiple organisms, scientists also are investigating the proteins produced by these genes.

Genes Chromosome (code for amino acids) Amino acids (join together DNA to form proteins) Ribosome (translates Nucleus mRNA to Cell (contains amino acids) Protein genome)

Figure 18 The central dogma is that the information in genes flows from DNA to RNA and RNA to proteins.

Genes are the primary information storage units, whereas proteins are the machines of a cell. Recall that when a gene is expressed, a protein is produced, as illustrated in **Figure 18**. Therefore, an understanding of how proteins function also is important. For instance, if the genome represents the words in a dictionary, the proteome, which represents all the proteins found in a cell, provides the definition of these words and how to use these words in a sentence. The large-scale study and cataloging of the structure and function of proteins in the human body is called **proteomics**. Proteomics allows researchers to look at hundreds or thousands of proteins at the same time. This type of broad analysis will better define both normal and disease states. Scientists anticipate that proteomics will revolutionize the development of new drugs to treat diseases such as Type II diabetes, obesity, and atherosclerosis.



What's BIOLOGY Got To Do With It?

Section 3 Assessment

Section Summary

- Researchers who worked on the HGP sequenced all nucleotides in the human genome.
- DNA fingerprinting can be used to identify individuals.
- DNA microarrays allow researchers to study all the genes in the genome simultaneously.
- Gene therapy might be used in the future to correct genetic disorders.
- Genomics is the study of an organism's genome and proteomics is the study of the proteins in the human body.

Understand Main Ideas

- 1. Relate the human genome to blueprints for a house.
- 2. Analyze the role of DNA fingerprinting in criminal investigations.
- 3. Indicate why the HapMap project is useful in diagnosing human disease.
- **4. Explain** the process of gene therapy. What is the ultimate goal of gene therapy?

Think Critically

5. Hypothesize Most of the human genome consists of noncoding DNA. Where did all of this noncoding DNA originate?

(MATHIN) Biology

6. If 1.5 percent of the human genome consists of protein-coding sequences, and the entire genome has 3.2×10^9 nucleotides, how many codons are in the human genome? Remember that a codon is three nucleotides in length.

In the Field

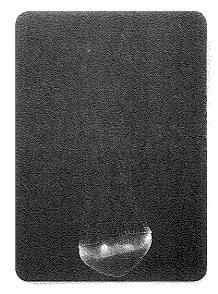
Career: Biomedical Research

Illuminating Medical Research

Have you ever watched fireflies glow on a summer evening? A chemical reaction in firefly cells produces light through a process called bioluminescence. Many marine organisms, like the jellyfish shown in the image, are also bioluminescent. The jellyfish species *Aequorea victoria* has emerged as a hero to biomedical researchers. This jellyfish produces a substance called green fluorescent protein (GFP), which makes parts of its body shine with an emerald green light.

Shining Light on Cell Functions Found off the west coast of North America, the diminutive Aequorea victoria is only five to ten centimeters in diameter. Its cells contain aequorin, a bioluminescent protein that emits a deep blue light. GFP absorbs this light and converts it into a glowing emerald green. In the early 1990s, scientists removed the GFP gene from Aequorea victoria and cloned it. Today, biomedical researchers can fuse GFP to other proteins inside cells of living organisms. When illuminated with light of a specific frequency, these marked proteins glow, making it possible to observe their behavior during cell processes.

Biological Marking at Work GFP allows scientists to determine where proteins are located during different stages of a cell's life, and to observe how proteins interact to produce disease. Researchers can attach GFP to a virus and observe the spread of the virus throughout the host.



Green fluorescent protein was first observed in bioluminescent jellyfishes.

By injecting tumor cells marked with GFP, scientists can analyze how they develop, spread, and destroy healthy cells over time.

Bioluminescent imaging can be used to evaluate the effectiveness of various treatments on these types of tumors. Ultimately, scientists hope to incorporate GFP directly into human tumor cells, then use bioluminescence to identify the mass as a separate cell population within the body. Easily differentiated from healthy cells and tissues, the glowing cancerous cells would be marked for treatment.

WRITING in Biology

Research and Communicate GFP is used to investigate the effectiveness of gene therapy, vaccines, and cancer treatments. Research how GFP is used in cancer studies and share your findings with classmates.



- E

38.FF

198/01

letter.

OCCUPANT

BIOLAB

FORENSICS: HOW CAN GENETIC ENGINEERING BE USED TO SOLVE A CRIME?

Background: Although all humans are similar genetically, variations do occur in certain segments of DNA. When cut with restriction enzymes, the variety of sizes of these fragments can be used to determine the source of a sample of DNA. In this lab, DNA from suspects will be analyzed.

Question: Based on the DNA samples, were any of the suspects at the scene?

Materials

various DNA samples
electrophoresis chamber
power source
micropipette and tips
prepared agarose gels
restriction enzyme
microcentrifuge tubes and rack
sample-loading dye
nontoxic dye
staining and destaining containers
DNA fragments of known size (control)
ruler
ice in foam container
water bath at 37°C

Safety Precautions

Procedure

- 1. Read and complete the lab safety form.
- 2. Read the entire procedure.
- 3. Label your DNA samples.
- **4.** Design and construct a data table you can use to record your observations when you perform gel electrophoresis of your samples.
- **5.** Your teacher will instruct you how to prepare your samples, set up the gel electrophoresis equipment, load your samples, and run the electrophoresis.

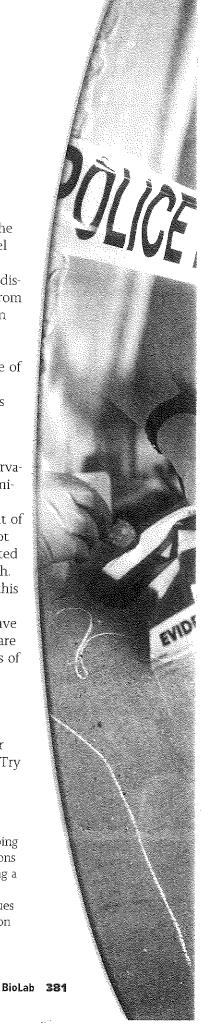
- **6.** Use the gel-staining dye to detect the location of DNA fragments in the gel for each of your samples.
- 7. Use a ruler to measure (in mm) the distance of each migrated DNA band from the wells. Record this information in your table.
- 8. Cleanup and Disposal Wash and return all reusable materials. Dispose of gels and other reagents in properly labeled containers. Wash your hands thoroughly.

Analyze and Conclude

- 1. Interpret Data Based on your observations, predict which suspect is incriminated by the DNA evidence.
- 2. Think Critically While the amount of DNA needed for electrophoresis is not large, the amount that can be extracted from a few hairs might not be enough. How might forensic scientists solve this problem?
- **3. Error Analysis** DNA fingerprints have a very high level of accuracy if they are run correctly. What are some sources of error that could lead to inaccurate results?
- **4. Plan Ahead** Suggest ways that you could improve your procedure and methods to avoid the sources of error listed in your answer for question 3. Try out your plans.

(WRITING in Biology

Plan a procedure. Find a news article describing the use of DNA fingerprinting in investigations such as a criminal investigation or identifying a bacterium involved in a disease outbreak. Write a mock lab that explains the techniques and steps that might be taken in the situation described by the article.





Study Guide Chapter

THEME FOCUS Patterns The genetics of each organism is the basis of diversity within species.

Genetic technology improves human health and quality of life.

Section 1 Applied Genetics

selective breeding (p. 360) inbreeding (p. 361) test cross (p. 362)

Selective breeding is used to produce organisms with desired traits.

- · Selective breeding is used to produce organisms with traits that are considered desirable.
- Hybridization produces organisms with the desired traits from parent organisms with different traits.
- Inbreeding creates pure breeds.
- A test cross can be used to determine an organism's genotype.

Section 2. DNA Testinology

genetic engineering (p. 363) genome (p. 364) restriction enzyme (p. 364) gel electrophoresis (p. 365) recombinant DNA (p. 366) plasmid (p. 366) DNA ligase (p. 366) transformation (p. 367) cloning (p. 367) polymerase chain reaction (p. 368) transgenic organism (p. 370)

Researchers use genetic engineering to manipulate DNA.

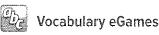
- Genetic engineering is used to produce organisms that are useful to humans.
- Recombinant DNA technology is used to study individual genes.
- DNA fragments can be separated using gel electrophoresis.
- · Clones can be produced by transforming bacteria with recombinant
- The polymerase chain reaction is used to make copies of small DNA sequences.
- Transgenic organisms are being created to increase the quality of human life.

Section 3 The Human Genome

DNA fingerprinting (p. 373) bioinformatics (p. 375) DNA microarray (p. 375) single nucleotide polymorphism (p. 376) haplotype (p. 378) pharmacogenomics (p. 378) gene therapy (p. 378) genomics (p. 378) proteomics (p. 379)

Genomes contain all of the information needed for an organism to grow and survive.

- Researchers who worked on the HGP sequenced all nucleotides in the human genome.
- DNA fingerprinting can be used to identify individuals.
- DNA microarrays allow researchers to study all the genes in the genome simultaneously.
- Gene therapy might be used in the future to correct genetic disorders.
- Genomics is the study of an organism's genome and proteomics is the study of the proteins in the human body.



Section 1

Vocabulary Review

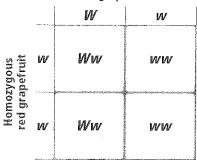
Fill in the blanks with the correct term from the Study Guide page.

- 1. A ______ is used to determine the genotype of a plant or animal.
- **2.** The offspring produced by _____ are homozygous for most traits.

Understand Main Ideas

Use the illustration below to answer questions 3 and 4.

Heterozygous white grapefruit



- **3.** What is the genotypic ratio of the offspring in the cross above?
 - A. 1:2:1
- C. All are homozygous recessive.
- **B.** 1:1
- D. All are heterozygous.
- **4.** The cross above could be used to determine the genotype of a parent with a dominant phenotype. What is this type of cross called?
 - A. a homozygous cross C. a test cross
 - B. a heterozygous cross D. a parental cross

Constructed Response

- 5. **THEME FOCUS Patterns** Predict the phenotype of the parent plants of hybrid tomato plants that grow fast and are resistant to pesticides. Explain.
- **6. Short Answer** How do polygenic traits affect selective breeding?
- 7. Discuss the advantages and disadvantages of selective breeding.

Think Critically

- **8. Explain** why purebred animals do not exist in the wild.
- 9. Determine Suppose a phenotype is controlled by more than one gene. Can a test cross be used to determine the genotype? Why or why not?

Section 2

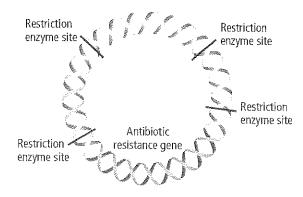
Vocabulary Review

Fill in the blank with the correct vocabulary term from the Study Guide page.

- 10. Transgenic animals are produced by _____
- 11. Biologists use ______ to join two DNA molecules together.
- **12.** During ______, a cell takes in DNA from outside the cell.
- **13.** Small, circular DNA molecules that are found in bacterial cells are called _____.

Understand Main Ideas

Use the illustration below to answer question 14.



- **14.** What is the role of the molecule above in DNA cloning?
 - A. to carry the foreign DNA into the host cell
 - B. to identify the source of DNA as foreign
 - **C.** to identify the host cell that has taken up the gene of interest
 - **D.** to make the foreign DNA susceptible to digestion with enzymes



Chapter 1 Assessment

15. Based on the sequences below, which enzyme produces a blunt end? The cut site is indicated by the *.

C*GGCC G A. Eagl G CCGG°C B. EcoRV GAT*ATC CTA*TAG C. NsiI A TGCA*T T"ACGT A T°CG A D. Tagl

16. Why is the polymerase chain reaction used?

A GC*T

A. to amplify DNA

C. to ligate DNA

B. to cut DNA

D. to separate DNA

Constructed Response

- 17. Predict what effect genetic engineering will have on the evolution of a species.
- 18. Short Answer Suppose you transform bacteria with a recombinant DNA plasmid and by mistake grow the transformed cells without an antibiotic. What result would you observe? Why?
- 19. Interpret the Figure Refer to Figure 9 to make a flowchart diagramming the steps in the PCR.

Think Critically

20. Conclude A recombinant DNA molecule was created by joining a plasmid vector and a DNA fragment. Gel electrophoresis verified that the plasmid and the DNA fragment ligated.



- a. Which lane in the gel corresponds to the recombinant DNA?
- **b.** Which lane corresponds to the plasmid?
- c. Which lane represents cleaving using a restriction enzyme of the recombinant DNA molecule?

21. Differentiate The plasmid below was cut to produce the five fragments shown in the diagram. The fragments then were separated by gel electrophoresis. Draw a diagram of a gel and the location of each fragment. Label ends as positive or negative.

1633 base pairs (bp)
3444443343344344444444444444444444444
257 bp
7) (1) (3) (3)
1108 bp
A(t)(t)(t)(t)(t)(t)(t)(t)(t)(t)(t)
1400 bp
$\chi(H1)(\phi(H1)(H1)(H1)(H1)(H1)(H1)(H1)_{H1}$
601 bp
A1841 (1914) (1914) (1914) (1914)

22. Assess A small DNA molecule was cleaved with several different restriction enzymes, and the size of each fragment was determined by gel electrophoresis. The following data were obtained.

DNA Fragmentati HindIII	on Patterns Created	by <i>EcoR</i> I and
Enzyme	Number of Fragments	Fragment Size (kilobases)
EcoRi	2	1.5 kb 1.5 kb
HindIII	1	3.0 kb
EcoRI + HindIII	3	0.8 kb 0.7 kb
		1.5 kb

- a. Is the original DNA linear or circular?
- **b.** Draw a restriction-site map showing distances consistent with the data.

Section 3

Vocabulary Review

Fill in the blanks with the correct vocabulary term from the Study Guide page.

- 23. The field of _____ uses computers to index and organize information created by sequencing the human genome.
- 24. Genetic variations that are located close together are called



Understand Main Ideas

- **25.** Which statement about the human genome is false?
 - A. The human genome contains approximately 25,000 genes.
 - **B.** The human genome contains long stretches of DNA with no known function.
 - **C.** The human genome was sequenced by scientists from around the world.
 - **D.** The human genome contains nucleotide sequences that all code for proteins.
- **26.** What are variations in specific nucleotides that are linked to human diseases called?
 - A. proteomes
 - B. haplotypes
 - C. single nucleotide polymorphisms
 - D. genomes
- 27. For what purpose is DNA fingerprinting used?
 - A. to sequence DNA from bacteria
 - B. to separate DNA fragments
 - **C.** to identify individuals who have committed crimes
 - **D.** to identify single nucleotide polymorphisms

Constructed Response

- **28. Short Answer** Discuss the advantages and disadvantages of using DNA microarrays.
- **29. Short Answer** List three ways patients will benefit from pharmacogenomics.
- **30.** What impact does sequencing the human genome have on diagnosing and treating diseases?

Think Critically

- **31. Describe** how DNA microarrays and DNA sequencing can be used to identify a defective gene.
- 32. ARRENS IN MINIMAN A forensic scientist finds a strand of hair at a crime scene. Draw a flowchart and explain the steps that the forensic scientist has to take to determine the identity of the person to whom the hair belongs.

Summative Assessment

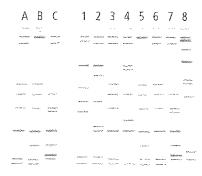
- 33. Explain the importance of the completion of the human genome project. What are some health discoveries that are a result of this project?
- 34. WRITINGIA Biology Write a paragraph discussing the approach you would take to create a transgenic organism and the drawbacks to creating it.

Document-Based Questions

The data below were obtained during a study on mosquito biting patterns. DNA fingerprints were obtained from individuals A, B, and C who were bitten by mosquitoes. In order to determine which mosquitoes bit each individual, a group of mosquitoes was collected and their DNA fingerprints were obtained. The mosquitoes were numbered 1–8.

Use the data to answer the questions below.

Data obtained from: Michael, et al. 2001. Quantifying mosquito biting patterns on humans by DNA fingerprinting of blood meals. *American Journal of Tropical Medicine and Hygiene* 65(6): 722–728.



- **35.** Examine the banding patterns and match each individual with the mosquito(es) that bit him or her.
- **36.** What can researchers gain by knowing which mosquito bit which individual?
- **37.** Based on your answer to question 35, what is a disadvantage of using this DNA fingerprinting to identify disease-carrying mosquitoes in the environment?

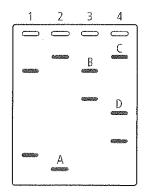
Standardized Test Practice

Cumulative

Multiple Choice

- 1. Which describes the process of cytokinesis?
 - A. chromosomes duplicate
 - B. spindle disintegrates
 - C. nucleus disappears
 - D. cytoplasm divides

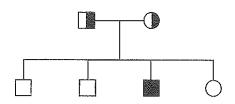
Use the illustration below to answer questions 2 and 3.



- 2. The figure above shows bands of DNA that were separated using gel electrophoresis. Which band contains the smallest DNA fragments?
 - A. Band A
 - B. Band B
 - C. Band C
 - D. Band D
- **3.** What could the results of this gel electrophoresis show to a scientist?
 - A. the amount of noncoding DNA present
 - B. the fingerprint of a person's DNA
 - C. the number of genes in a piece of DNA
 - D. the random patterns of DNA
- **4.** Which process plays a part in genetic recombination?
 - A. asexual reproduction
 - **B.** cytokinesis
 - C. independent assortment
 - **D.** mitotic division

- 5. Which correctly lists the following terms in order from smallest to largest: DNA, chromatin, chromosomes, nucleosomes?
 - A. chromatin, chromosomes, DNA, nucleosomes
 - B. chromosomes, DNA, chromatin, nucleosomes
 - C. DNA, nucleosomes, chromatin, chromosomes
 - D. nucleosomes, DNA, chromatin, chromosomes

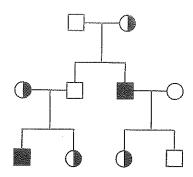
Use the figure below to answer question 6.



- **6.** In a particular family, one child out of four is born with Tay-Sachs disease. Which pair of symbols represents the parents of these offspring?
 - A. ____
- 7. Which is a stop codon in mRNA?
 - A. AUG
 - B. AUU
 - C. CAU
 - D. UAA
- **8.** In a triploid organism, how many alleles are present for each gene per cell?
 - A. 1
 - **B.** 3
 - **C**. 6
 - D. 9

Shork Answer

Use the figure below to answer question 9.



- 9. The pedigree in the figure tracks a recessive, sex-linked genetic disease. Explain the meaning of the symbols in the last generation.
- 10. Why are the protein-coding regions of most human genomes identical?
- 11. If hemophilia is a sex-linked recessive gene, what is the chance that a father with hemophilia and a mother who is a carrier for hemophilia will have a boy with hemophilia? Explain.
- 12. Compare and contrast the two major processes in protein synthesis.
- 13. List three genetic disorders; classify them as dominant or recessive; and name the affected organ systems.
- 14. Why might it take many generations to develop a purebred animal?
- 15. List the purine bases and the pyrimidine bases in DNA; explain their împortance in DNA structure.

Externed Response

16. Give the names of two DNA mutations, and illustrate how each one would change the following DNA sequence.

CGATTGACGTTTTAGGAT

- 17. Chemosynthetic autotrophs might have evolved long before the photosynthetic ones that currently are more common on Earth. Propose an explanation for this difference in evolution.
- 18. Explain how the noncoding sequences in the human genome make it difficult to interpret the DNA code.
- 19. Even though chloroplasts and mitochondria perform different functions, their structures are similar. Relate the similarity of their structures to their functions.

Essay Ouestion

Suppose a scientist uses gel electrophoresis to separate the DNA extracted from a cell line. After performing the experiment, the scientist observes that several bands are missing and that other bands have traveled to the far end of the gel.

Using the information in the paragraph above, answer the following question in essay format.

20. Using what you know about DNA separation and gel electrophoresis, explain what might have gone wrong with the experiment. Then, describe how to adjust the experimental procedures to test your explanation.

NEED EXTRA HELP?																				
If You Missed Question	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20
Review Section	9.2	13.2	13.3	10.3	12.1	11.1	12.3	10.3	11.1	13.3	11.3	12.3	11.2	13.1	12.1	12.4	2.2	13.3	7.3	13.2