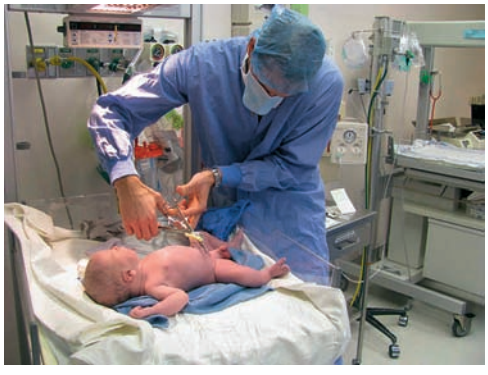
CHAPTER 11

# Applications of Biotechnology



## CHAPTER OUTLINE



## Thinking of Preserving Baby's Cells?

*Banking on future medical treatments—skepticism required.*

How young can you be to donate blood? Normally, a baby's umbilical cord is discarded after birth. However, blood that remains in the cord contains stem cells that can be collected and preserved in hopes that it may be useful in the future. Stem cells have the ability to develop into any of your cells. Would you like to have some of your child's embryonic stem cells preserved so that they might be used to cure illness or repair injury? If the child experiences tissue or organ problems due to damage, disease, age, or genetic defects, these preserved cells might be used to generate tissues to repair or replace the damage. It is thought that these stem cells have the potential to be cloned and used to treat such conditions as: cancer, brain injury, juvenile diabetes, renal failure and spinal cord injuries. The cost of private cord blood banking is about $2,000 for collection and $125 per year for storage.

While at first glance this sounds to be "the way to go" in assuring that your child's future health problems may be dealt with efficiently, the procedure is controversial. Even though public cord blood banking is supported by the medical community, the American Academy of Pediatrics 2007 Policy Statement on Cord Blood Banking noted that physicians should be aware of unsubstantiated claims of private cord blood banks. Other aspects of this controversy center on issues and such facts as:

- ✓ The likelihood of using your own stem cells is 1 in 435.
- ✓ The European Union Group on Ethics states the legitimacy of commercial cord blood banks for such use should be questioned because they sell a service that presently has no real therapeutic value.
- ✓ Cord blood cells have the same genes as the donor and cannot be used to treat genetic diseases of the donor.

- **What are stem cells?**
- **What does it mean to clone cells?**
- **Would you buy into a cord blood donation program?**

## Background Check

**Concepts you should already know to get the most out of this chapter:**

- All organisms use the same genetic code to make proteins (chapter 8)
- DNA codes for genetic information that codes for the cell's proteins (chapter 8)
- Proteins influence how the organism or the cell looks, behaves, and functions (chapter 10)

# 11.1 Why Biotechnology Works

The discovery of DNA's structure in 1953 opened the door to a new era of scientific investigation. **Biotechnology** is a collection of techniques that provide the ability to manipulate the genetic information of an organism *directly*. As a result, scientists can accomplish tasks that were not feasible just 60 years ago. The field of biotechnology has enabled scientists to produce drugs more cheaply than before; to correct genetic mutations; to create cells that are able to break down toxins and pollutants in the environment; and to develop more productive livestock and crops. Biotechnology promises more advances in the near future.

The key to understanding biotechnology is understanding the significant role that DNA plays in determining the genetic characteristics of an organism. In the cell's nucleus, chromosomes are made of DNA and histone proteins. The genetic information for the cell is the sequence of nucleotides that make up the DNA molecule. Genes are regions of the DNA's nucleotide sequence that contain the information to direct the synthesis of specific proteins. In turn, these proteins produce the characteristics of the cell and organism when the gene is expressed by transcription and translation.

DNA in nucleus → proteins in cells → phenotype of organism

The nearly universal connection among DNA, protein expression, and the organism's phenotype is central to biotechnology. If an organism has a unique set of phenotypes, it has a unique set of DNA sequences. The more closely related organisms are, the more similar are their DNA sequences.

### 11.1 CONCEPT REVIEW

1. Why is the word *directly* so important to the understanding of the definition of biotechnology?
2. Why can DNA in one organism be used to make the same protein in another organism?

# 11.2 Comparing DNA

It is useful to distinguish between individual organisms on the basis of their DNA. Comparisons of DNA can be accomplished in two general ways. Both rely on the fact that genetically different organisms will have different nucleotide sequences in their DNA. The two methods are DNA fingerprinting and DNA sequencing. DNA fingerprinting looks at patterns in specific portions of the DNA of an organism. DNA sequencing looks directly at the nucleotide sequence.

Because both of these approaches have advantages and disadvantages, scientists choose between them depending on their needs. DNA fingerprinting allows for a relatively quick look at larger areas of the organism's genetic information. It is useful to distinguish between organisms—such as possible suspects in a court trial. DNA sequencing creates a very detailed look at a relatively small region of the organism's genetic information. DNA sequencing is the most detailed look that we are able to have of the organism's genetic information.

## DNA Fingerprinting

**DNA fingerprinting** is a technique that uniquely identifies individuals on the basis of short pieces of DNA. Because no two people have the same nucleotide sequences, they do not generate the same lengths of DNA fragments when their DNA is cut with enzymes. Even looking at the many pieces of DNA that are produced in this manner is too complex. Therefore, scientists don't look at all the possible fragments but, rather, focus on differences found in pieces of DNA that form repeating patterns in the DNA. By focusing on these regions with repeating nucleotide sequences, it is possible to determine whether samples from two individuals have the same number of repeating segments (Outlooks 11.1).

### DNA Fingerprinting Techniques

In the scenario presented in Outlooks 11.1, a crime was committed and the scientists had evidence in the form of body fluids from the criminal. These body fluids contained cells with the criminal's DNA. The DNA in these cells was used as a template to produce enough DNA for analysis. The **polymerase chain reaction (PCR)** is a technique used to generate large quantities of DNA from small amounts (How Science Works 11.1).

Using PCR and the suspect's DNA, scientists were able to replicate regions of human DNA that are known to vary from individual to individual. This created large quantities of DNA so that DNA fingerprinting could be performed. Scientists target areas of the suspect's DNA that contains *variable number tandem repeats*. **Variable number tandem repeats (VNTRs)** are sequences of DNA that are repeated a variable number of times from one individual to another. For example, in a given region of DNA, one person may have a DNA sequence

## OUTLOOKS 11.1

# The First Use of a DNA Fingerprint in a Criminal Case

In 1988, a baker in England was the first person in the world to be convicted of a crime on the basis of DNA evidence. Colin Pitchfork's crime was the rape and murder of two girls. The first murder occurred in 1983. The initial evidence in this case consisted of the culprit's body fluids, which contained his proteins and DNA. On the basis of the proteins, the police were able to create a molecular description of the culprit. The problem was that this description matched 10% of the males in the local population, and the police were unable to identify just one person. In 1986, there was another murder that closely matched the details of the 1983 killing. Another male, Richard Buckland, was the prime suspect for the second murder. In fact, while being questioned, Buckland admitted to the most recent killing but had no knowledge of the first killing. The clues still did not point consistently to a single person.

Meanwhile, the scientists at a nearby university had been working on a new forensic technique—DNA fingerprinting.

To track down the killer, police asked local men to donate blood or saliva samples. Between 4,000 and 5,000 local men participated in the dragnet. None of the volunteers matched the culprit's DNA. Interestingly, Buckland's DNA did not match the culprit's DNA, either. He was later released because his confession was false. It wasn't until after someone reported that Colin Pitchfork had asked a friend to donate a sample for him and offered to pay several others to do the same that police arrested Pitchfork. Pitchfork's DNA matched that of the killer's.

This is a good example of how biotechnology helps the search for truth within the justice system. The additional evidence from DNA was able to provide key information to identify the culprit.

repeated 4 times, whereas another may have the same sequence repeated 20 times (figure 11.1).

Once enough DNA was generated through PCR, the DNA needed to be treated so that the VNTRs would be detectable. To detect the varying number of VNTRs, the replicated DNA sample is cut into smaller pieces with **restriction enzymes**. **Restriction sites** are DNA nucleotide sequences that attract restriction enzymes. When the restriction enzymes bind to a restriction site, the enzyme cuts the DNA molecule into two molecules. **Restriction fragments** are the smaller DNA fragments that are generated after the restriction enzyme has cut the selected DNA into smaller pieces. Some of the fragments of DNA that are generated by restriction enzymes will contain the regions with VNTRs. The fragments with VNTRs will vary in size from person to person because some individuals have more repeats than others. Restriction enzymes are used to create fragments of DNA that might be different from one individual to the next.

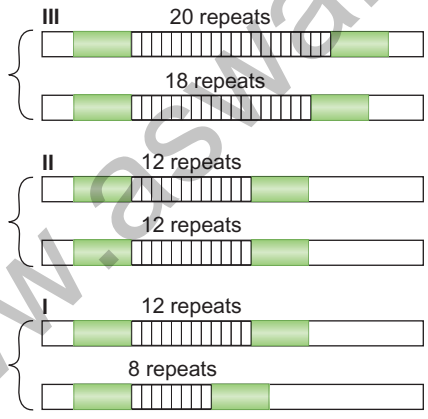



**FIGURE 11.1 Variable Number Tandem Repeats**
Variable number tandem repeats (VNTRs) are short sequences of DNA that are repeated often. The repeated sequences are attached end-to-end. This illustration shows the VNTRs for three individuals. The individual in I has 8 repeats on 1 chromosome and 12 on the homologous chromosome. They are heterozygous. The individual in II is homozygous for 12 repeats. The individual in III is heterozygous for a different number of repeats—18 and 20.

In DNA fingerprinting, scientists look for different lengths of restriction fragments as an indicator of differences in VNTRs.

**Electrophoresis** is a technique that separates DNA fragments on the basis of size (How Science Works 11.2). The shorter DNA molecules migrate more quickly than the long molecules. As differently sized molecules are separated, a banding pattern is generated. Each band is a differently sized restriction fragment. Each person's unique DNA banding pattern is called a DNA fingerprint (figure 11.2). The process of DNA fingerprinting includes the following basic stages:

1. DNA is obtained from a source, which may be as small as one cell.
2. PCR is used to make many copies of portions of the DNA that contain VNTRs.
3. Restriction enzymes are used to cut the VNTR DNA into pieces so that the VNTRs can be detected.
4. To detect the differences in the VNTRs, the pieces are separated by electrophoresis.
5. Comparisons between patterns can be made.

### DNA Fingerprinting Applications

With DNA fingerprinting, the more similar the banding patterns are from two different samples, the more likely the two samples are from the same person. The less similar the patterns, the less likely the two samples are from the

# HOW SCIENCE WORKS 11.1

## Polymerase Chain Reaction

Polymerase chain reaction (PCR) is a laboratory procedure for copying selected segments of DNA from larger DNA molecules. With PCR, a single cell can provide enough DNA for analysis and identification. Scientists start with a sample of DNA that contains the desired DNA region. The types of samples that can be used include semen, hair, blood, bacteria, protozoa, viruses, mummified tissues, and frozen cells. Targeting specific portions of DNA for replication enables biochemists to manipulate DNA more easily. When many copies of this DNA have been produced it is easy to find, recognize, and manipulate.

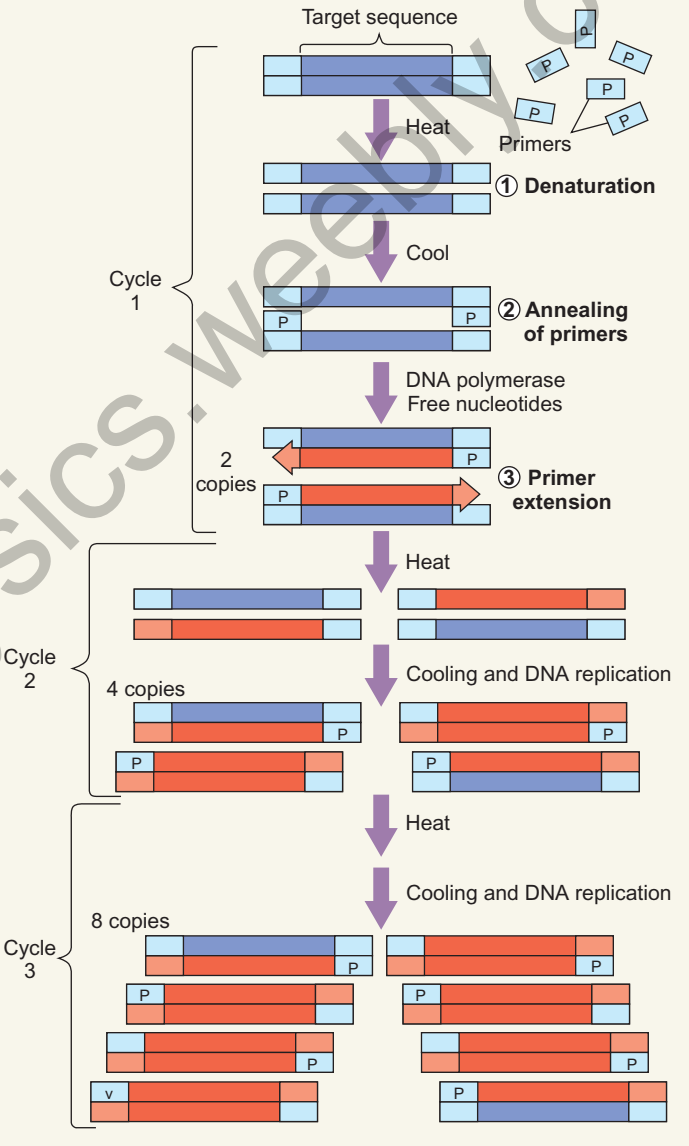
PCR is a test-tube version of the cellular DNA replication process and requires similar components. The DNA from the sample specimen serves as the template for replication. Free DNA nucleotides are used to assemble new strands of DNA. DNA polymerase, which has been purified from bacteria cells, is used to catalyze the PCR reaction.

DNA primers are short stretches of single-stranded DNA, which are used to direct the DNA polymerase to replicate only certain regions of the template DNA. These primer molecules are specifically designed to flank the ends of the target region's DNA sequence and point the DNA polymerase to the region between the primers. The PCR reaction is carried out by heating the target DNA, so that the two strands of DNA fall away from each other. This process is called *denaturation*. Once the nitrogenous bases on the target sequence are exposed and the reaction cools, the primers are able to attach to the template molecule. The primers *anneal* to the template. The primers *anneal* (that is, stick or attach) to the template. The primers are able to target a particular area of DNA because the primer nucleotide sequence pairs with the template DNA sequence using the base-pairing rules.

Purified DNA polymerase is the enzyme that drives the DNA replication process. The presence of the primer, attached to the DNA template and added nucleotides, serves as the substrate for the DNA polymerase. Once added, the polymerase extends the DNA molecule from the primer down the length of the DNA. Extension continues until the polymerase falls off of the template DNA. The enzyme incorporates the new DNA nucleotides in the growing DNA strand. It stops when it reaches the other end, having produced a new copy of the target sequence.

The elegance of PCR is that it allows the exponential replication of DNA. Exponential, or logarithmic, growth is a doubling in number with each round of PCR. With just one copy of template DNA, there will be a total of two copies at the end of one replication cycle. During the second round, both copies are used as a template. At the end of the second round, there is a total of 4 copies. The number of copies of the target DNA increases very quickly. With each round of replication, the number doubles—8, 16, 32, 64. Each round of replication takes only minutes. Thirty rounds of replication in PCR can be performed within 2.5 hours. Starting with just one copy of DNA and 30 rounds of replication, it is possible to produce over half a billion copies of the desired DNA segment.

Because this technique can create useful amounts of DNA from very limited amounts, it is a very sensitive test for the presence of specific DNA sequences. Frequently, the presence of a DNA sequence indicates the presence of an infectious agent or a disease-causing condition.




**PCR Replication**

During cycle one of PCR, the template DNA is denatured, so that the two strands of DNA separate. This allows the primers to attach (anneal) to the template DNA. DNA polymerase and DNA nucleotides, which are present for the reaction, create DNA by extending from the primers. During cycle 2, the same process occurs again, but the previous round of replication has made more template available for further replication. Each subsequent cycle essentially doubles the amount of DNA.

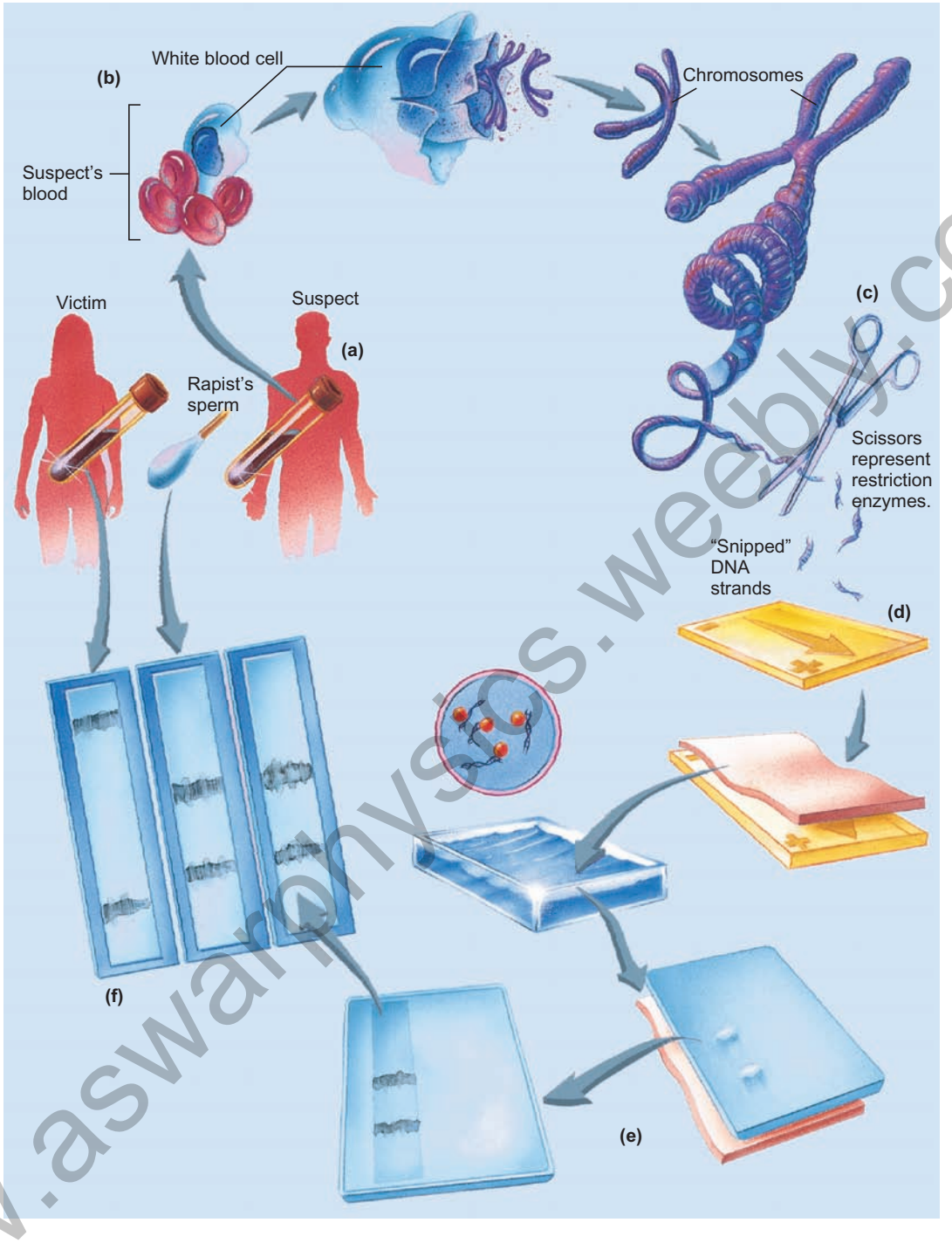



**FIGURE 11.2 DNA Fingerprints**
*(a)* Because every person's DNA is unique, *(b)* when samples of an individual's DNA are collected and subjected to restriction enzymes, the cuts occur in different places and DNA fragments of different sizes result. *(c)* Restriction enzymes can cut DNA at places where specific sequences of nucleotides occur. *(d)* When the cut DNA fragments are separated by electrophoresis, *(e)* the smaller fragments migrate more quickly than the larger fragments. This produces a pattern, called a DNA fingerprint, that is unique and identifies the person who provided the DNA. *(f)* The victim's DNA is on the left. The rapist's DNA is in the middle. The suspect's DNA is on the right. The match in banding patterns between the suspect and the rapist indicates that they are the same.

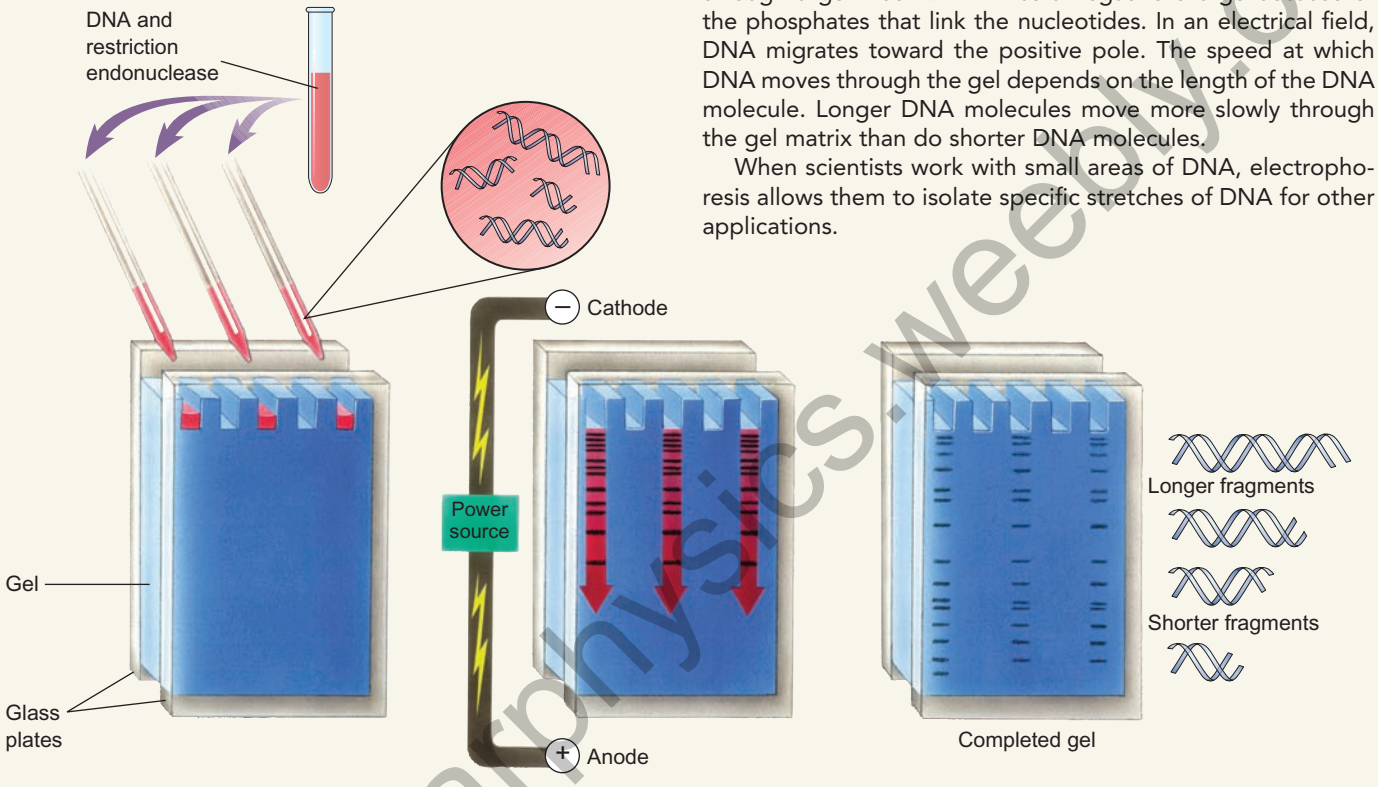
## HOW SCIENCE WORKS 11.2

### Electrophoresis

Electrophoresis is a technique used to separate molecules, such as nucleic acids, proteins, or carbohydrates. Electrophoresis separates nucleic acids on the basis of size. DNA is too long for scientists to work with when taken directly from the cell. To make the DNA more manageable, scientists cut the DNA into smaller pieces. Restriction enzymes are frequently used to cut large DNA molecules into smaller pieces. After the DNA is broken into smaller pieces, electrophoresis is used to separate differently sized DNA fragments.

Electrophoresis uses an electric current to move DNA through a gel matrix. DNA has a negative charge because of the phosphates that link the nucleotides. In an electrical field, DNA migrates toward the positive pole. The speed at which DNA moves through the gel depends on the length of the DNA molecule. Longer DNA molecules move more slowly through the gel matrix than do shorter DNA molecules.

When scientists work with small areas of DNA, electrophoresis allows them to isolate specific stretches of DNA for other applications.




Mixture of DNA fragments of different sizes in solution placed at the top of "lanes" in the gel

Electric current applied; fragments migrate down the gel by size—smaller ones move faster (and therefore go farther) than larger ones

same person. In criminal cases, DNA samples from the crime site can be compared with those taken from suspects. If 100% of the banding pattern matches, it is highly probable that the suspect was at the scene of the crime and is the guilty party. The same procedure can be used to confirm a person's identity, as in cases of amnesia, murder, or accidental death.

DNA fingerprinting can be used in paternity cases that determine the biological father of a child. A child's DNA is a unique combination of both the mother's DNA and the father's DNA. The child's DNA fingerprint is unique, but all the bands in the child's DNA fingerprint should be found in either the mother's or the father's fingerprint. To determine paternity, the child's DNA, the mother's DNA, and DNA from the man who is alleged to be the father are collected.

The DNA from all three is subjected to PCR, restriction enzymes, and electrophoresis. During analysis of the banding patterns, scientists account for the child's banding pattern by linking each DNA band to a DNA band of the mother and the presumed father. Bands that are common to both the biological mother and the child are identified and eliminated from further consideration. If all the remaining bands can be matched to the presumed father, it is extremely likely that he is the father (figure 11.3). If there are bands that do not match the presumed father's, then there are one

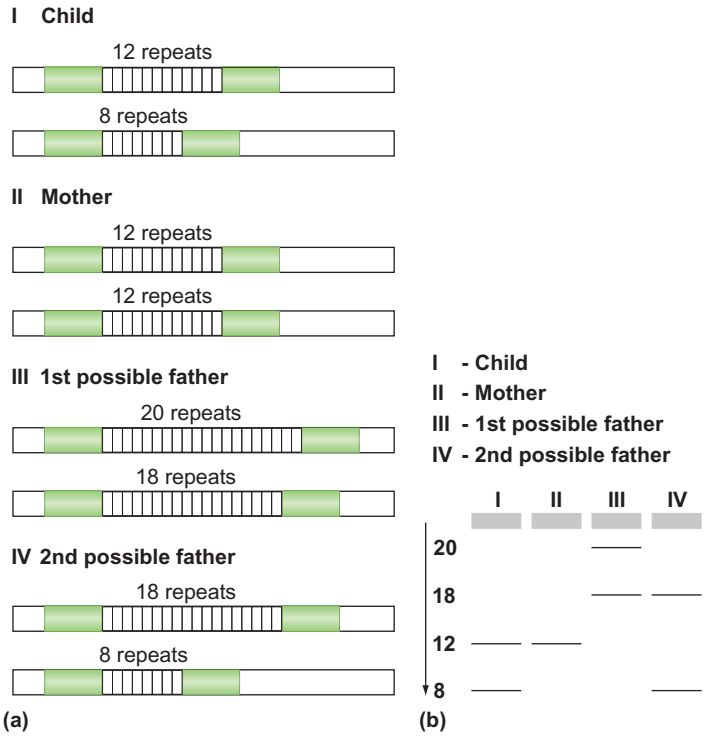



**FIGURE 11.3 Paternity Determination**
*(a)* This illustration shows the VNTRs for four different individuals—a child, the mother, one possible father, and a second possible father. *(b)* Using PCR, electrophoresis, and DNA fingerprinting analysis, it is possible to identify the child's father. The mother possesses the "12" band and has passed that to her child. The mother did not give the child the child's "8" band because the mother does not have an "8" band herself. The child's "8" band must have come from the father. Of the two men under consideration, only man IV has the "8" band, so man IV is the father. Now stop for a moment and think about the principles of genetics. If man IV is the father, why doesn't the child have an "18" band?

of two conclusions: (1) The presumed father is not the child's biological father, or (2) the child has a new mutation that accounts for the unique band. This last possibility can usually be ruled out by considering multiple regions of DNA, because it is extremely unlikely that the child will have multiple new mutations.

## Gene Sequencing and the Human Genome Project

The Human Genome Project (HGP) was a 13-year effort to determine the human DNA sequence. Work began in 1990. It was first proposed in 1986 by the U.S. Department of Energy (DOE) and was cosponsored soon after by the National Institutes of Health (NIH). These agencies were the main research agencies within the U.S. government responsible for developing and planning the project. Estimates are that the United States spent over $3 billion on the Human Genome Project.

Many countries contributed both funds and labor resources to the Human Genome Project. At least 17 countries other than the United States participated, including Australia, Brazil, Canada, China, Denmark, France, Germany, Israel, Italy, Japan, Korea, Mexico, the Netherlands, Russia, Sweden, and the United Kingdom. The Human Genome Project was one of the most ambitious projects ever undertaken in the biological sciences.

The data that these countries produced are stored in powerful computers, so that the information can be shared. To get an idea of the size of this project, consider that a human Y chromosome (one of the smallest of the human chromosomes) is composed of nearly 60 million paired nucleotides. The larger X chromosome may be composed of 150 million paired nucleotides. The entire human genome consists of 3.12 billion paired nucleotides. That is roughly the same number as all the letter characters found in about 2,000 copies of this textbook.

### Human Genome Project Techniques

Two kinds of work progressed simultaneously to determine the sequence of the human genome. First, *physical maps* were constructed by determining the location of specific "markers" and the proximity of these markers to genes. The markers were known sequences of DNA that could be located on the chromosome. This physical map was used to organize the vast amount of data produced by the second technique, which was for the labs to determine the exact order of nitrogenous bases of the DNA for each chromosome. Techniques exist for determining base sequences (How Science Works 11.3). The challenge is storing and organizing the information from these experiments, so that the data can be used.

A slightly different approach was adopted by Celera Genomics, a private U.S. corporation. Although Celera Genomics started later than the labs funded by the Department of Energy and National Institute of Health it was able to catch up and completed its sequencing at almost the same time as the government-sponsored programs by developing new techniques. Celera jumped directly to determining the DNA sequence of small pieces of DNA without the physical map. It then used computers to compare and contrast the short sequences, so that it could put them together and assemble the longer sequence. The benefit of having these two organizations as competitors was that, when they finished their research, they could compare and contrast results. Amazingly, the discrepancies between their findings were declared insignificant.

### Human Genome Project Applications

The first draft of the human genome was completed early in 2003, when the complete nucleotide sequence of all 23 pairs of human chromosomes was determined. By sequencing the human genome, it is as if we have now identified all the words in the human gene "dictionary." Continued analysis will provide the definitions for these words—what these words tell the cell to do.

The information provided by the human genome project is extremely useful in diagnosing diseases and providing genetic counseling to those considering having children. This

# HOW SCIENCE WORKS 11.3

## DNA Sequencing

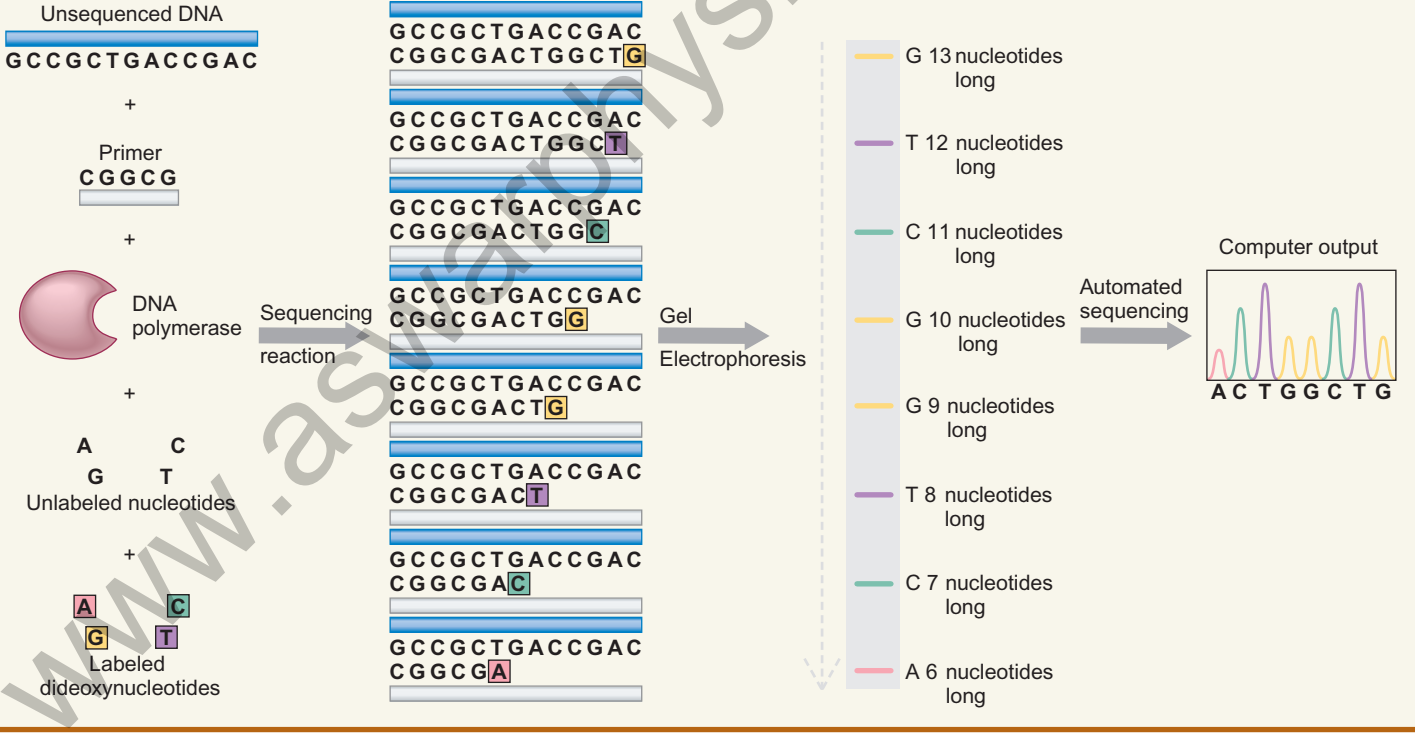
DNA sequencing uses electrophoresis to separate DNA fragments of different lengths. A DNA synthesis reaction is set up that includes DNA from the region being investigated. The reaction also includes (1) DNA polymerase, (2) a specific DNA primer, (3) all DNA nucleotides (G, A, T, and C), and (4) a small amount of 4 kinds of chemically altered DNA nucleotides. DNA polymerase is the enzyme that synthesizes DNA in cells by using DNA nucleotides as a substrate. The DNA primer gives the DNA polymerase a single place to start the DNA synthesis reaction. All of these components work together to allow DNA synthesis in a manner very similar to cellular DNA replication.

The DNA sequencing process also adds nucleotides that have been chemically altered in two ways: (1) The altered nucleotides are called dideoxyribonucleosides because they contain a dideoxyribose sugar rather than the normal deoxyribose. Dideoxyribose has one less oxygen in its structure than deoxyribose. (2) The four kinds of dideoxyribonucleotides (A, T, G, C) are each labeled with a different flourescent dye so that each of the four nucleotides is colored differently. During DNA sequencing, the DNA polymerase randomly incorporates either a normal DNA nucleotide or a dideoxyribonucleotide. When the dideoxyribonucleoside is used, two things happen: (1) No more nucleotides can be added to the DNA strand, and (2) the DNA strand is now tagged with the fluorescent label of the dideoxynucleotide that was just incorporated.

As a group, the DNA molecules that are created by this technique have the following properties:

1. They all start at the same point, because they all started with the same primer.
2. There are copies of DNA molecules that had their replication halted at each nucleotide in the sequence of the sample DNA when a dideoxyribose nucleotide was incorporated.
3. DNA molecules of the same length (number of nucleotides) are labeled with the same color of fluorescent dye.

Electrophoresis separates this collection of molecules by size, because the shortest DNA molecules move fastest. The DNA sequence is determined by reading the color sequence from the shortest DNA molecules to the longest DNA molecules. The pattern of colors matches the order of the nucleotides in the DNA. The color pattern that is generated by the sequencing gel is the order of the nucleotides. Automated sequencing is done by using a laser beam to read the colored bands. A printout is provided as peaks of color to show the order of the nucleotides.



information can identify human genes and proteins that can be targets for drugs and new gene therapies. Once it is known where an abnormal gene is located and how it differs in base sequence from the normal DNA sequence, steps could be taken to correct the abnormality. Further defining the human genome will also result in the discovery of new families of proteins and will help explain basic physiological and cell biological processes common to many organisms. All this information will increase the breadth and depth of the understanding of basic biology.

It was originally estimated that there were between 100,000 and 140,000 genes in the human genome, because scientists were able to detect so many different proteins. DNA sequencing data indicate that there are only about 20,000 protein-coding genes—only about twice as many as in a worm or a fly. Our genes are able to generate several different proteins per gene because of alternative splicing (figure 11.4). Alternative splicing occurs much more frequently than previously expected. Knowing this information provides insights into the evolution of humans and will make future efforts to work with the genome through bioengineering much easier.

There is a concern that, as our genetic makeup becomes easier to determine, some people may attempt to use this information for profit or political power. Consider that some health insurance companies refuse to insure people with "preexisting conditions" or those at "genetic risk" for certain abnormalities. Refusing to provide coverage would save these companies the expense of future medical bills incurred by "less than perfect" people. While this might be good for insurance companies, it raises major social questions about fair and equal treatment and discrimination.

Another fear is that attempts may be made to "breed out" certain genes and people from the human population to create a "perfect race." Intentions such as these superficially appear to have good intentions, but historically they have been used by many groups to justify discrimination against groups of individuals or even to commit genocide.

## Other Genomes

While some scientists refine our understanding of the human genome, others are sequencing the genomes of other organisms. Representatives of each major grouping of organisms have been investigated, and the DNA sequence data have been made available to the general public through a centralized government website. This centralized database has made the exchange and analysis of scientific information easier than ever (table 11.1). The information gained from these studies

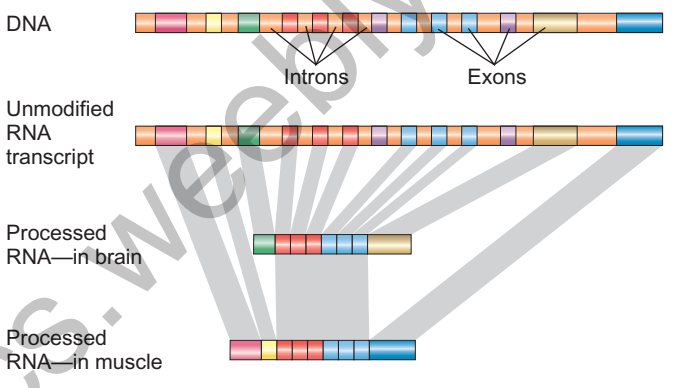



**FIGURE 11.4 Different Proteins—One Gene**
This illustration shows a stretch of DNA that contains a gene. Protein-coding regions (exons) of this gene are shown in different colors. Introns that do not code for protein and are not transcribed into RNA are shown in a single color—rust. Alternative splicing allows different tissue to use the same gene but make slightly different proteins. The gray bands show how some exons are used to form both proteins, whereas other exons are used on only one protein.

**TABLE 11.1 Completed and Current Genome Projects**

The Human Genome Project has sparked major interest in nonhuman genomes. The investigation of some genomes has been very organized. Other investigations have been less directed, whereby only sequences of certain regions of interest have been reported. Regardless, information on many genomes is available at the National Center for Biotechnology Information website.

| Taxonomic Group | Genome Examples | Number of Different Genomes Represented |
|---|---|---|
| Viruses and retroviruses and bacteriophages | Herpes virus, human papillomavirus, HIV | Over 1,560 |
| Bacteria | Anthrax species, *Chlamydia* species, *Escherichia coli, Pseudomonas* species, *Salmonella* species | Over 200 |
| Archaea | *Halobacterium* species, *Methanococcus* species, *Pyrococcus* species, *Thermococcus* species | Over 21 |
| Protists | *Cryptosporidium* species, *Entamoeba histolytica, Plasmodium* species | Over 45 |
| Fungi | Yeast, *Aspergillus, Candida* | Over 70 |
| Plants | Thale cress *(Arabidopsis thaliana)*, tomato, lotus, rice | Over 20 |
| Animals | Bee, cat, chicken, chimp, cow, dog, frog, fruit fly, mosquito, nematode, pig, rat, sea urchin, sheep, zebra fish | Over 100 |
| Cellular organelles | Mitochondria, chloroplasts | |

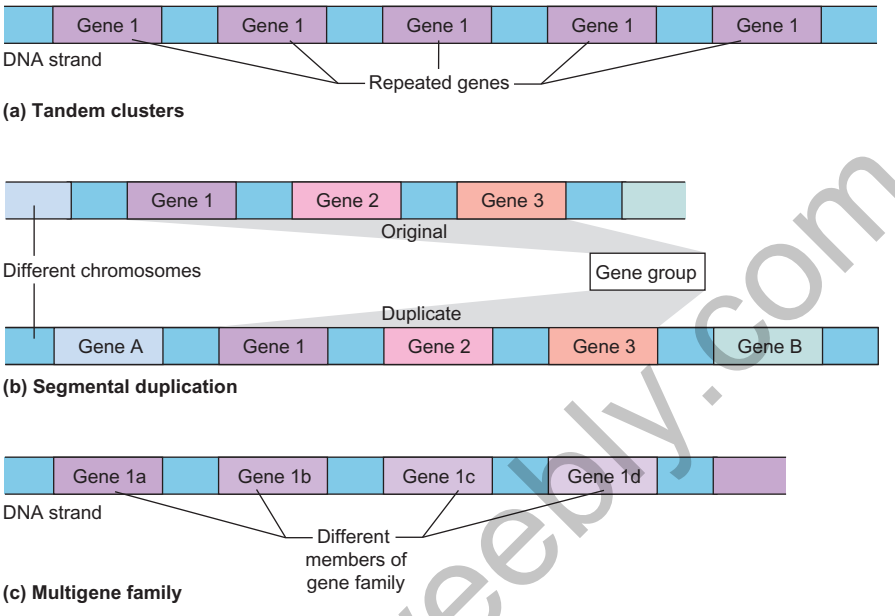



**FIGURE 11.5 Patterns in Protein Coding Sequences**
*(a)* Tandem clusters are identical or nearly identical repeats of one gene. *(b)* Segmental duplications are duplications of sets of genes. These may occur on the same chromosome or different chromosomes. *(c)* Multigene families are repeats of similar genes. The genes are similar because regions are conserved from gene to gene, but many regions have changed significantly.

might lead to new treatments for disease and a better understanding of evolutionary relationships.

## Patterns in Protein-Coding Sequences

As scientists sequenced the human genome and compared it with other genomes, certain patterns became apparent.

**Tandem clusters** are grouped copies of the same gene that are found on the same chromosome (figure 11.5). For example, the DNA that codes for ribosomal RNA is present in many copies in the human genome. From an evolutionary perspective, the advantage to the cell is the ability to create large amounts of gene product quickly from the genes found in tandem clusters.

**Segmental duplications** are groups of genes that are copied from 1 chromosome and moved as a set to another chromosome. These types of gene duplications allow for genetic backups of information. If either copy is mutated, the remaining copy can still provide the necessary gene product sufficient for the organism to live. Because the function of the mutated copy of the gene is being carried out by the normal gene, the mutated copy may take on new function if it accumulates additional mutations. This can allow evolution to occur more quickly (figure 11.5b).

**Multigene families** are groups of different genes that are closely related. When members of multigene families are closely inspected, it is clear that certain regions of the genes carry similar nucleotide sequences. Hemoglobin is a member of the globin gene family. There are several different hemoglobin genes in the human genome. Evolutionary patterns can be tracked at the molecular level by examining gene families across species. The portions of genes that show very little change across many species represent portions of the protein that are important for function. Scientists reason that regions that are important for function will be intolerant of change and stay unaltered over time. Again, using hemoglobin as an example, it is possible to compare the hemoglobin genes of different organisms to identify specific changes in the gene. Such comparisons can lead to a better understanding of how organisms are related to each other evolutionarily (figure 11.5c).

The following are a few more interesting facts obtained by comparing genomes:

- Eukaryotic genomes are more complex than prokaryotic genomes. Eukaryotic genomes are, on average, nearly twice the size of prokaryotic genomes. Eukaryotic genomes devote more DNA to regulating gene expression. Only 1% of human DNA actually codes for protein.
- The number of genes in a genome is not a reflection of the size or complexity of an organism. Humans possess roughly 21,000 genes. Roundworms have about 26,000 genes, and rice plants possess 32,000 to 55,000 genes.
- Eukaryotes create multiple proteins from their genes because of alternative splicing. Prokaryotes do not. Nearly 25% of human DNA consists of intron sequences, which are removed during splicing. On average, each human gene makes between 4.5 and 5 different proteins because of alternative splicing.
- There are numerous, virtually identical genes found in very distantly related organisms—for example, mice, humans, and yeasts.
- Hundreds of genes found in humans and other eukaryotic organisms appear to have resulted from the transfer of genes from bacteria to eukaryotes at some point in eukaryotic evolution.
- Chimpanzees have 98–99% of the same DNA sequence as humans. All the human "races" are about 99.9% identical at the DNA level. In fact, there is virtually no scientific reason for the concept of "race," because the amount of variation *within* a race is as great as the amount of variation *between* races.
- Genes are unequally distributed between chromosomes and unequally distributed along the length of a chromosome.

### Patterns in Non-Coding Sequence

Protein-coding DNA is not the only reason for examining DNA sequences. The regions of DNA that do not code for protein are more important than once thought. Many non-coding sequences are involved with the regulation of gene expression.

A recent and more accurate map of the human genome from Britain focuses on *copy number variations,* or CNVs. These are segments in the genetic code that can be deleted or copied; most are deletions and a small number are duplications. The new map has also revealed that humans have:

1. 75 "jumping genes," or *transposable elements* (regions of the genetic code that can move from one place to another in the genome of an individual).
2. more than 250 genes that can lose one of the two copies in chromosomes and not causing any obvious consequences, and
3. 56 genes that can join together, potentially forming new genes.

### New Fields of Knowledge

The ability to make comparisons of the DNA of organisms has led to the development of three new fields in biology—*genomics, transcriptomics,* and *proteomics*. **Genomics** is the comparison of the genomes of different organisms to identify similarities and differences. Species relatedness and gene similarities can be determined from these studies. When the DNA sequence of a gene is known, **transcriptomics** looks at when, where, and how much mRNA is expressed from a gene. Finally, **proteomics** examines the proteins that are predicted from the DNA sequence. From these types of studies, scientists are able to identify gene families that can be used to determine how humans have evolved at a molecular level. They can also examine how genes are used in an organism throughout its body and over its life span. They can also better understand how a protein works by identifying common themes from one protein to the next.

### 11.2 CONCEPT REVIEW

3. What types of questions can be answered by comparing the DNA of two different organisms?
4. What techniques do scientists use to compare DNA?
5. What benefits does the Human Genome Project offer?
6. What is the purpose of the PCR?
7. What role does electrophoresis play in DNA comparisons?
8. What are tandem clusters, segmental duplications and multigene families?

## 11.3 The Genetic Modification of Organisms

For thousands of years, civilizations have attempted to improve the quality of their livestock and crops. Cows that produce more milk or more tender meat were valued over those that produced little milk or had tough meat. Initial attempts to develop improved agricultural stocks were limited to selective breeding programs, in which only the organisms with the desired characteristics were allowed to breed. As scientists asked more sophisticated questions about genetic systems, they developed ways to create and study mutations.

Although this approach was a very informative way to learn about the genetics of an organism, it lacked the ability to create a specific desired change. Creating mutations is a very haphazard process. However, today the results are achieved in a much more directed manner using biotechnology's ability to transfer DNA from one organism to another. **Transformation** takes place when a cell gains new genetic information from its environment. Once new DNA sequences are transferred into a host cell, the cell is genetically altered and begins to read the new DNA and produce new cell products, such as enzymes. The resulting new form of DNA is called **recombinant DNA.**

A **clone** is an exact copy of biological entities, such as genes, organisms, or cells. The term refers to the outcome, not the way the results are achieved. Many whole organisms "clone" themselves simply by how they reproduce; bacteria divide by cell division and produce two genetically identical cells. Strawberry plants clone themselves by sending out runners and establishing new plants. Many varieties of fruit trees and other plants are cloned by making cuttings of the plant and rooting the cuttings. With the development of advanced biotechnology techniques, it is now possible to clone specific genes from an organism. It is possible to put that cloned gene into the cell of an entirely different species.

### Genetically Modified Organisms

**Genetically modified** (**GM**) organisms contain recombinant DNA. Viruses, bacteria, fungi, plants, and animals are examples of organisms that have been engineered so that they contain genes from at least one unrelated organism.

As this highly sophisticated procedure has been refined, it has become possible to splice genes quickly and accurately from a variety of species into host bacteria or other host cells by a process called *gene cloning* (How Science Works 11.4). Genetically modified organisms are capable of expressing the protein-coding regions found on recombinant DNA. Thus, the organisms with the recombinant DNA can make products they were previously unable to make. Since they can rapidly reproduce to large numbers, industrial-sized cultures

# HOW SCIENCE WORKS 11.4

## Cloning Genes

Cloning a specific gene begins with cutting the source DNA into smaller, manageable pieces with restriction enzymes. Next, there are several basic steps that occur in the transfer of DNA from one organism to another:

1. The source DNA is cut into a usable size by using restriction enzymes.

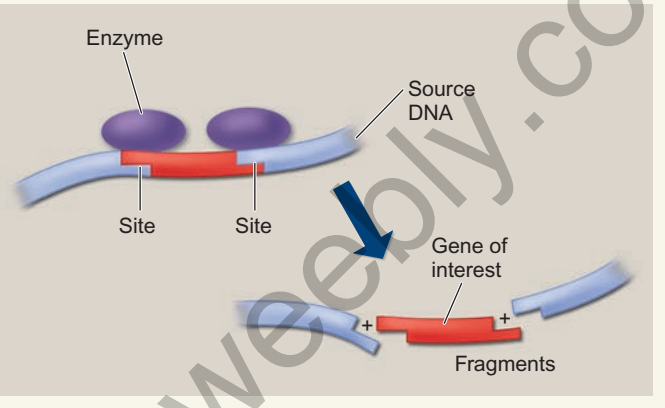
   The source DNA is usually isolated from a large number of cells. Therefore, It consists of many copies of an organism's genome. The source DNA is cut into many small fragments with restriction enzymes. Isolating the small portion of DNA that contains the gene of interest can be difficult because the gene of interest is found on only a few of these fragments. To identify the desired fragments, scientists must search the entire collection. The search involves several steps.
2. The DNA fragments are attached to a carrier DNA molecule.

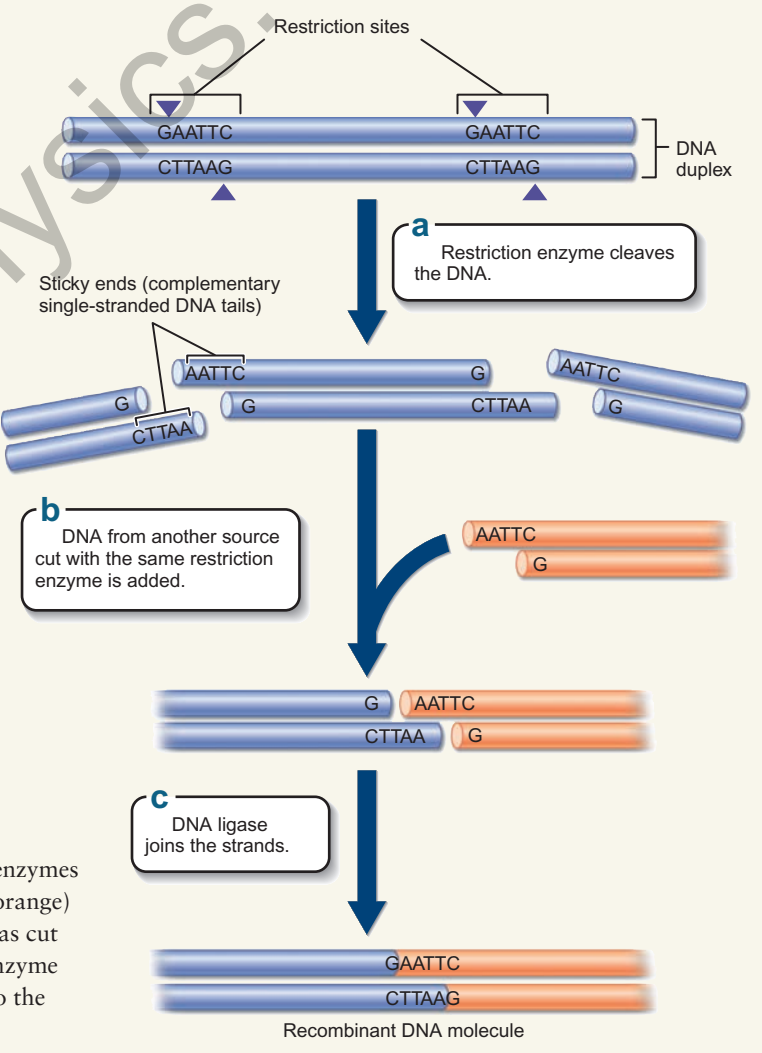
   The first step is to attach every fragment of source DNA to a carrier DNA molecule. A **vector** is the term scientists use to describe a carrier DNA molecule. Vectors usually contain special DNA sequences that facilitate attachment to the fragments of source DNA. Vectors also contain sequences that promote DNA replication and gene expression.

   A plasmid is one example of a vector that is used to carry DNA into bacterial cells. A **plasmid** is a circular piece of DNA that is found free in the cytoplasm of some bacteria. Therefore, the plasmid must be cut with a restriction enzyme, so that the plasmid DNA will have sticky ends, which can attach to the source DNA. The enzyme ligase creates the covalent bonds between the plasmid DNA and the source DNA, so that a new plasmid ring is formed with the source DNA inserted into the ring. The plasmid and its inserted source DNA is recombinant DNA. Because there are many different source DNA fragments, this process results in many different plasmids, each with a different piece of source DNA. All of these recombinant DNA plasmids constitute a **DNA library** for the entire source genome.
3. The carrier DNA molecule, with its attached source DNA, is moved into an appropriate cell for the carrier DNA. In the cell, the new DNA is replicated or expressed.

### Cutting Genomic DNA

The first step in cloning a specific gene is to cut the source DNA into smaller, manageable pieces with restriction enzymes.



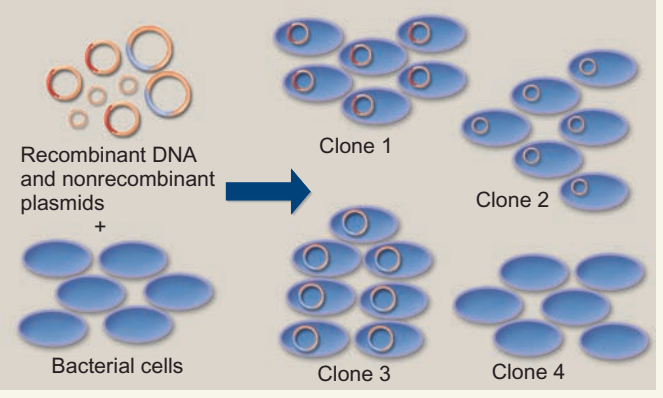

### Creating Recombinant DNA

The source DNA is cut with restriction enzymes to create sticky ends. The vector DNA (orange) has compatible sticky ends, because it was cut with the same restriction enzyme. The enzyme ligase is used to bond the source DNA to the vector DNA.

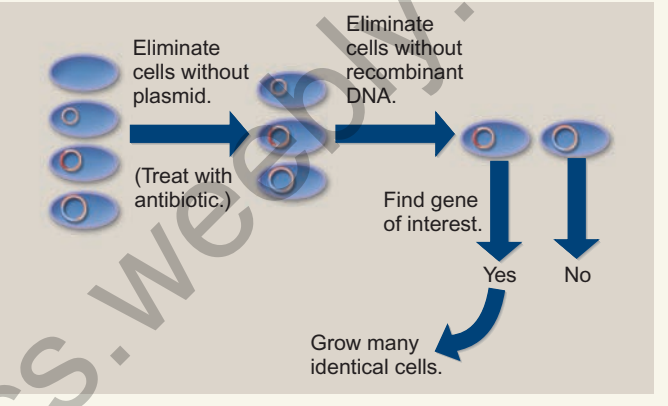
## HOW SCIENCE WORKS 11.4 (*Continued*)

The second step in the cloning process is to mix the DNA library with bacterial cells that will take up the DNA molecules. Transformation occurs when a cell gains genetic information from its environment. Each transformed bacterial cell carries a different portion of the source DNA from the DNA library. These cells can be grown and isolated from one another.

The third step is to screen the DNA library contained within the many different transformed bacterial cells to find those that contain the DNA fragment of interest. Once the bacterial cells with the desired recombinant DNA are identified, the selected cells can be reproduced and, in the process, the desired DNA is cloned.




**Transformation**
Bacterial cells pick up the plasmids with recombinant DNA and are transformed. Different cells pick up plasmids with different genomic DNA inserts.




**Screening the DNA Library**
A number of techniques are used to eliminate cells that do not carry plasmids with attached source DNA. Once these cells are eliminated from consideration, the remaining cells are screened to find those that contain the genes of interest.

of bacteria can synthesize large quantities of proteins. For example, recombinant DNA procedures are responsible for the production of:

- Human insulin, used in the control of diabetes (figure 11.6)
- Nutritionally enriched "golden rice," capable of supplying poor people in less developed nations with beta-carotene, which is missing from normal rice
- Interferon, used as an antiviral agent
- Human growth hormone, used to stimulate growth in children lacking this hormone
- Somatostatin, a brain hormone implicated in growth.

The primary application of GM technology is to put herbicide-resistance or pest-resistance genes into crop plants. Edible GM crops are used mainly for animal feed. In agricultural practice, two kinds of genetically modified organisms have received particular attention. One involves the insertion of genes from a specific kind of bacterium called *Bacillus thuringiensis israeliensis* (Bti). Bti produces a protein that causes the destruction

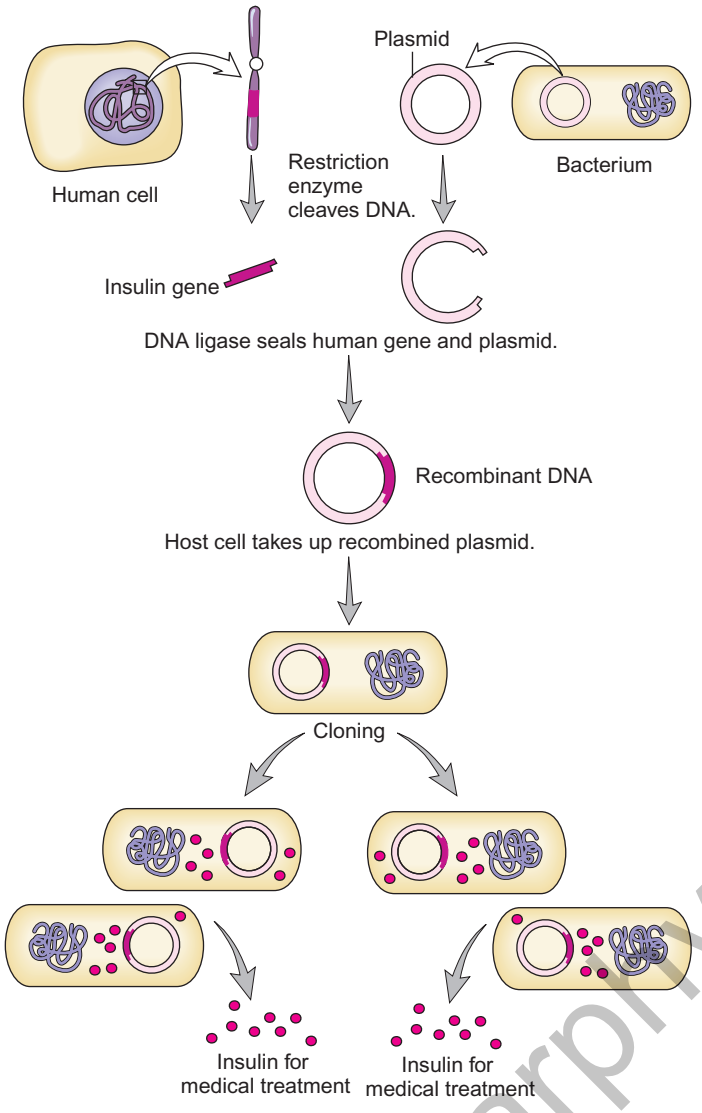



**FIGURE 11.6 Human Insulin from Bacteria**
The gene-cloning process is used to place a copy of the human insulin gene into a bacterial cell. As the bacterial cell reproduces, the human DNA it contains is replicated along with the bacterial DNA. The insulin gene is expressed along with the bacterial genes and the colony of bacteria produces insulin. This bacteria-produced human insulin is both more effective and cheaper than previous therapies, which involved obtaining insulin from the pancreas of slaughtered animals.


(a)


(b)

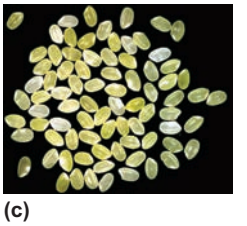

(c)

**FIGURE 11.7 Application of Genetically Modified Organisms**
Soybeans, corn, cotton, Hawaiian papaya, tomatoes, rapeseed, sugarcane, sugar beets, sweet corn, and rice are a short list of GM crops being grown and sold. *(a)* One of the most important applications of this technology involves the insertion of genes that make a crop plant resistant to herbicides. Therefore, the field can be sprayed with an herbicide and kill the weeds without harming the crop plant. *(b)* Normal rice does not produce significant amounts of beta-carotene. Beta-carotene is a yellow-orange compound needed in the diet to produce vitamin A. *(c)* Genetically modified "golden rice" can provide beta-carotene to populations that have no other sources of this nutrient.

of the lining of the gut of insects that eat it. It is a natural insecticide. To date, the gene has been inserted into the genetic makeup of several crop plants, including corn. In field tests, the genetically engineered corn was protected against some of its insect pests, but there was some concern that pollen grains from the corn might be blown to neighboring areas and affect nontarget insect populations. In particular, a study of monarch butterflies indicated that populations of butterflies adjacent to fields of this genetically engineered corn were negatively affected. One could argue that since the use of Bti corn results in less spraying of insecticides in cornfields, this is just a trade-off.

A second kind of genetically engineered plant involves inserting a gene for herbicide resistance into the genome of certain crop plants (figure 11.7a). The value of this to farmers is significant. For example, a farmer could plant cotton with very little preparation of the field to rid it of weeds. When both the cotton and the weeds begin to grow, the field could be sprayed with a specific herbicide that would kill the weeds but not harm the herbicide-resistant cotton. This has been field-tested and it works. Critics have warned that the genes possibly could escape from the crop plants and become part of the genome of the weeds that we are trying to control, thus creating "super-weeds."

Many more products have been manufactured using these methods. Genetically modified cells are not only used as factories to produce chemicals but also for their ability to break down many toxic chemicals. **Bioremediation** is the use of living organisms to remove toxic agents from the environment. There has been great success in using genetically modified bacteria to clean up oil spills and toxic waste dumps.

## Genetically Modified Foods

Although some chemicals have been produced in small amounts from genetically engineered microorganisms, crops such as turnips, rice, soybeans, potatoes, cotton, corn, and tobacco can generate tens or hundreds of kilograms of specialty chemicals per year. Such crops have the potential of supplying the essential amino acids, fatty acids, and other nutrients now lacking in the diets of people in underdeveloped and developing nations. Researchers have also shown, for example, that turnips can produce interferon (an antiviral agent), tobacco can create antibodies to fight human disease, oilseed rape plants can serve as a source of human brain hormones, and potatoes can synthesize human serum albumin that is indistinguishable from the genuine human blood protein (figure 11.7b and c).

Many GM crops also have increased nutritional value yet can be cultivated using traditional methods. There are many concerns regarding the development, growth, and use of GM foods. Although genetically modified foods are made of the same building blocks as any other type of food, the public is generally wary. Countries have refused entire shipments of GM foods that were targeted for hunger relief. However, we may eventually come to a point where we can no longer choose to avoid GM foods. As the world human population continues to grow, GM foods may be an important part of meeting the human population's need for food. The following are some of the questions being raised about genetically modified food:

- Is tampering with the genetic information of an organism ethical?
- Is someone or an agency monitoring these crops to determine if they are moving beyond their controlled ranges?
- What safety precautions should be exercised to avoid damaging the ecosystems in which GM crops are grown?
- What type of approval should these products require before they are sold to the public?
- Is it necessary to label these foods as genetically modified?

## Gene Therapy

The field of biotechnology allows scientists and medical doctors to work together and potentially cure genetic disorders. Unlike contagious diseases, genetic diseases cannot be transmitted, because they are caused by a genetic predisposition for a particular disorder—not separate, disease-causing organisms, such as bacteria and viruses. **Gene therapy** involves inserting genes, deleting genes, and manipulating the action of genes in order to cure or lessen the effect of genetic diseases. These therapies are very new and experimental. While these lines of investigation create hope, many problems must be addressed before gene therapy becomes a reliable treatment for many disorders.

The strategy for treating someone with gene therapy varies, depending on the disorder. When designing a gene therapy treatment, scientists have to ask exactly what the problem is. Is the mutant gene not working at all? Is it working normally but there is too little activity? Is there too much protein being made? Or is the gene acting in a unique, new manner? If there is no gene activity or too little gene activity, the scientists need to introduce a more active version of the gene. If there is too much activity or if the gene is engaging in a new activity, this excess activity must first be stopped and then the normal activity restored.

To stop a mutant gene from working, scientists must change it. This typically involves inserting a mutation into the protein-coding region of the gene or the region that is necessary to activate the gene. Scientists have used some types of viruses to do this in organisms other than humans. The difficulty in this technique is to mutate only that one gene without disturbing the other genes and creating more mutations in other genes. Developing reliable methods to accomplish this is a major focus of gene therapy. Once the mutant gene is silenced, the scientists begin the work of introducing a "good" copy of the gene. Again, there are many difficulties in this process:

- Scientists must find a way of returning the corrected DNA to the cell.
- The corrected DNA must be made a part of the cell's DNA, so that it is passed on with each cell division, it doesn't interfere with other genes, and it can be transcribed by the cell as needed (figure 11.8).
- Cells containing the corrected DNA must be reintroduced to the patient.

## The Cloning of Organisms

*Cloning* does not always refer to exchanging just a gene. Another type of cloning is the cloning of an entire organism. In this case, the goal is to create a new organism that is genetically identical to the previous organism. Cloning of multicellular organisms, such as Protists, plants, fungi and many kinds of invertebrate animals, often occur naturally during asexual reproduction and is duplicated easily in laboratories. The technique used to accomplish cloning in vertebrates is called *somatic cell nuclear transfer.* **Somatic cell nuclear transfer** removes a nucleus from a cell of the organism that will be cloned. After chemical treatment, that nucleus is placed into an egg cell that has had it original nucleus removed. The egg cell will use the new nucleus as genetic information. In successful cloning experiments with mammals, an electrical shock is used to stimulate the egg to begin to divide as if it were a normal embryo. After transferring the egg with its new nucleus into a uterus, the embryo grows normally. The resulting organism is genetically identical to the organism that donated the nucleus.

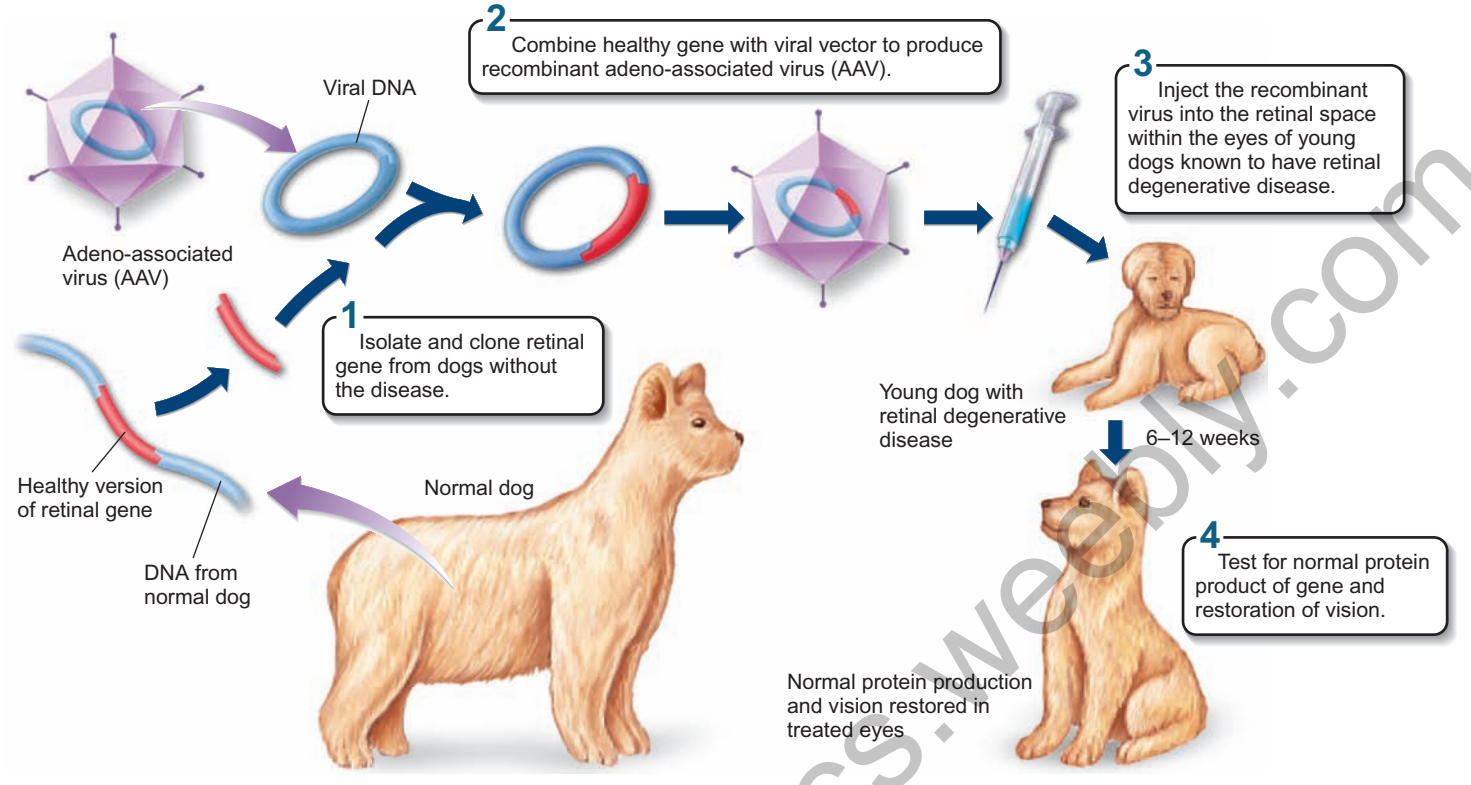



**FIGURE 11.8 Gene Therapy**
One method of introducing the correct genetic information to a cell is to use a virus as a vector. Here, a dog is treated for a degenerative disorder of the retina. The normal gene is spliced into the viral genome. The virus is then used to infect the defective retinal cells. When the virus infects the retinal cells, it carries the functional gene into the cell.

In 1996, a team of scientists from Scotland successfully carried out somatic cell nuclear transfer for the first time in sheep. The nucleus was taken from the mammary cell of an adult sheep. The embryo was transplanted into a female sheep's uterus, where it developed normally and was born (figure 11.9). This cloned offspring was named Dolly. This technique has been applied to many other animals, such as monkeys, goats, pigs, cows, mice, mules, and horses, and has been used successfully on humans. However, for ethical reasons, the human embryo was purposely created with a mutation that prevented the embryo from developing fully. The success rate of cloning animals is still very low for any animal, however; only 3–5% of the transplanted eggs develop into adults (figure 11.10).

A cloning experiment has great scientific importance, because it represents an advance in scientists' understanding of the processes of *determination* and *differentiation*. Recall that determination is the process a cell goes through to select which genes it will express. A differentiated cell has become a particular cell type because of the proteins that it expresses. Differentiation is more or less a permanent condition. The techniques that produced Dolly and other cloned animals use a differentiated cell and reverse the determination process, so that this cell is able to express all the genes necessary to create an entirely new organism. Until this point, scientists were not sure that this was possible.

### 11.3 CONCEPT REVIEW

9. A scientist can clone a gene. An organism can be a clone. How is the use of the word *clone* different in these instances? How is the use of the word *clone* the same in both uses?
10. What are some of the advantages of creating genetically modified (GM) foods? What are some of the concerns?
11. Describe how viruses are used in gene therapy.

## 11.4 Stem Cells

**Stem cells** are cells that are self-renewing and have not yet completed determination or differentiation, so they have the potential to develop into many different cell types. Scientists can generate stem cells by nuclear transfer techniques; they also occur naturally throughout the body. They are involved in many activities including tissue regeneration, wound healing, and cancer treatment.

If scientists had the ability to control differentiation it may allow the manipulation of an organism's cells or the insertion of cells into an organism to allow the regrowth of

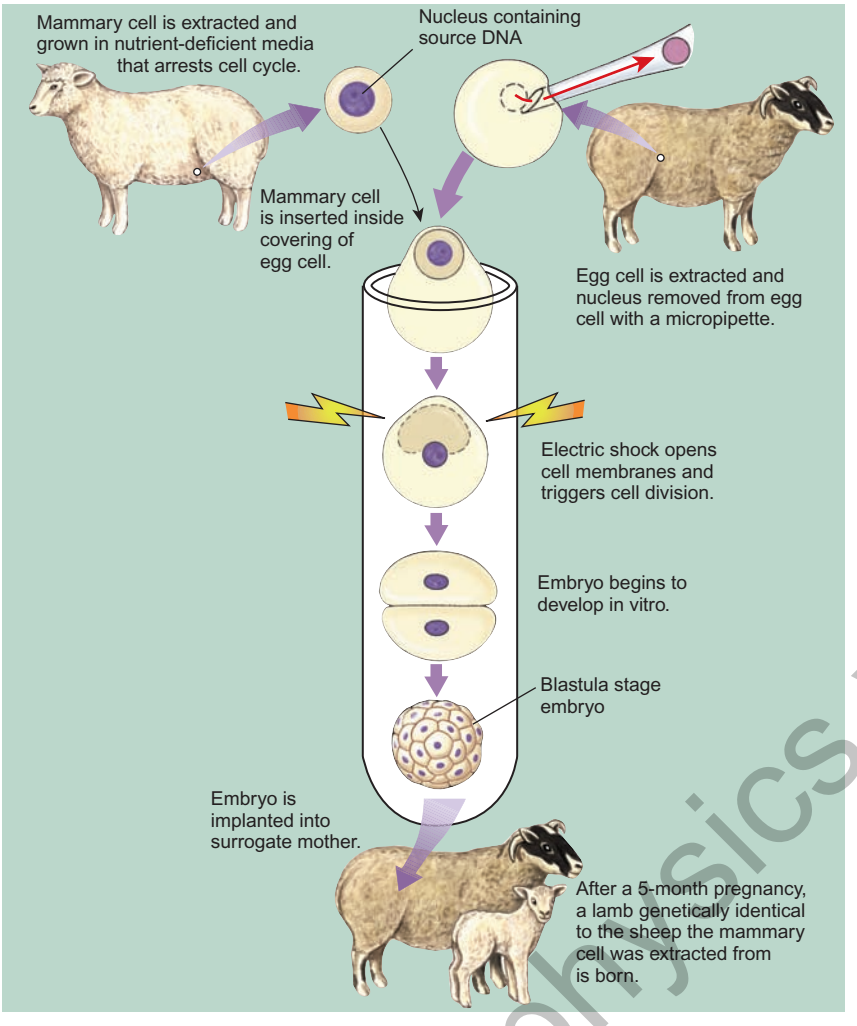



**FIGURE 11.9 Cloning an Organism**
The nucleus from the donor sheep is combined with an egg from another sheep. The egg's nucleus had previously been removed. The egg, with its new nucleus, is stimulated to grow by an electrical shock. After several cell divisions, the embryo is artificially implanted in the uterus of a sheep, which will carry the developing embryo to term.

(a) Copy Cat and her surrogate mother

(b) Rainbow the cat from which the nucleus to produce Copy Cat came.

**FIGURE 11.10 Success Rate in Cloning Cats**
Out of 87 implanted cloned embryos, CC (Copy Cat) is the only one to survive. This is comparable to the success rate in sheep, mice, cows, goats, and pigs. *(a)* Notice that CC is completely unlike her tabby surrogate mother. *(b)* "Rainbow" is her genetic donor, and both are female calico domestic shorthair cats.

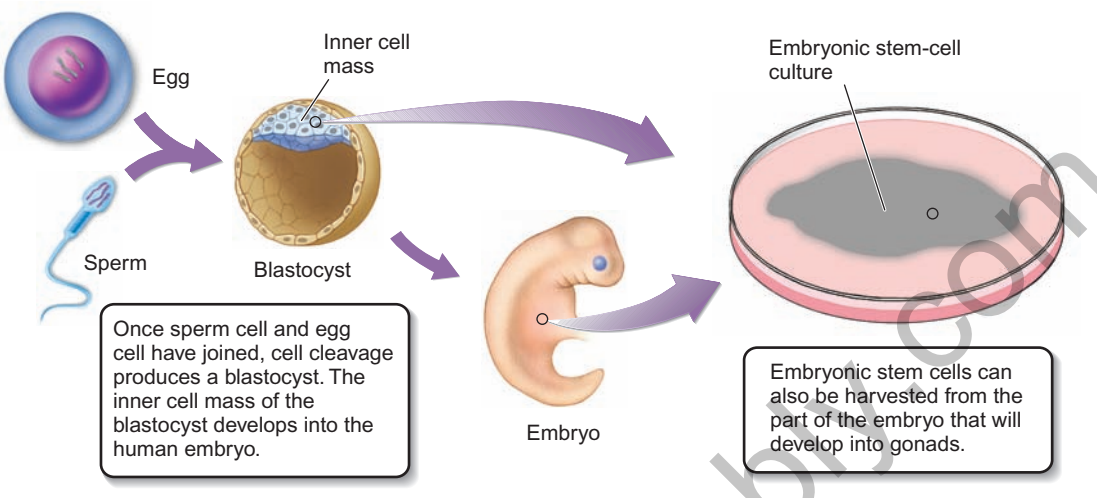



**FIGURE 11.11 The Culturing of Embryonic Stem Cells**
After fertilization of an egg with sperm, the cell begins to divide and form a mass of cells. Each of these cells has the potential to become any cell in the embryo. Embryonic stem cells may be harvested at this point or at other points in the determination process.

damaged tissues and organs in humans. This could aid in the cure or treatment of many medical problems, such as the repair of damaged knee cartilage, heart tissue from a heart attack, or damaged nerve tissue from spinal or head injuries. Some kinds of degenerative diseases occur because specific kinds of cells die or cease to function properly. Parkinson's disease results from malfunctioning brain cells, and many forms of diabetes are caused by malfunctioning cells in the pancreas. If stem cells could be used to replace these malfunctioning cells, normal function could be restored and the diseases cured.

## Embryonic and Adult Stem Cells

Because embryonic stem cells have not undergone determination and differentiation and have the ability to become *any* tissue in the body, they are of great interest to scientists. As an embryo develops, its stem cells go through the process of determination and differentiation to create all the necessary tissues. To study embryonic stem cells, scientists must remove them from embryos, destroying the embryos (figure 11.11).

Scientists have also explored other methods of obtaining stem cells. Embryonic stem cells reach an intermediary level of determination at which they are committed to becoming a particular *tissue* type, but not necessarily a particular *cell* type. An example of this intermediate determination occurs when stems cells become determined to be any one of several types of nerve cells but have not yet committed to becoming any one nerve cell. Scientists call these partially determined stem cells "tissue-specific." These types of stem cells can be found in adults. One example is hematopoietic stem cells. These cells are able to become the many different types of cells found in blood—red blood cells, white blood cells, and platelets (figure 11.12). The disadvantage of using these types of stem cells is that they have already become partially determined and do not have the potential to become every cell type.

## Personalized Stem Cell Lines

Scientists hope that eventually it will be possible to produce embryonic stem cells from somatic cells by using somatic cell nuclear transfer techniques similar to that used for cloning a sheep. This technique would involve transferring a nucleus from the patient's cell to a human egg that has had its original nucleus removed. The human egg would be allowed to grow and develop to produce embryonic stem cells. If the process of determination and differentiation can be controlled, new tissues, or even new organs, could be developed through what is termed *regenerative medicine*.

Under normal circumstances, organ transplant patients must always worry about rejecting their transplant and take strong immunosuppressant drugs to avoid organ rejection. Tissue and organs grown from customized stem cells would have the benefit of being immunologically compatible with the patient; thus, organ rejection would not be a concern (figure 11.13).

The potential therapeutic value of stem cells has resulted in the founding of many clinics around the world. These clinics offer stem cell-based therapies to patients with a variety of medical conditions. However the benefits of these therapies are as yet unproven and, it fact, have the potential for serious harm. "Stem cell tourism" is a phrase being used to describe this industry. Desperate patients travel to these clinics in hopes that such therapies will save their lives. This new industry is teeming with "medical tourist traps" offering unproven medical treatments to unsuspecting consumers. Unfortunately, the days of customized stem cells, stem cell therapies, and organ culture are still in the future.

### 11.4 CONCEPT REVIEW

12. Embryonic stem cells are found in embryos, and adult stem cells are found in adults. In what other ways are they different?
13. What benefits does stem cell research offer?
14. What are some of the concerns with research on stem cells?

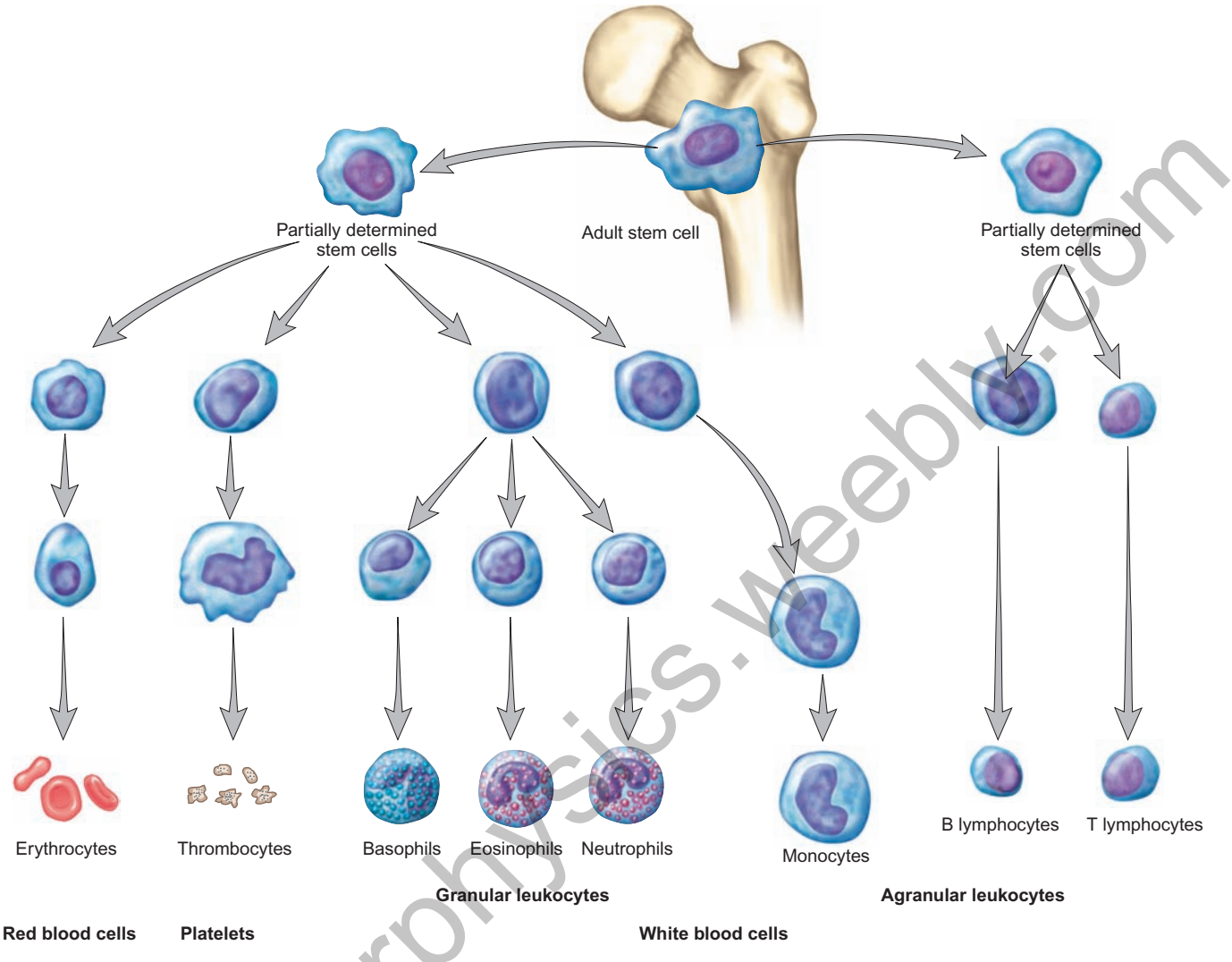



**FIGURE 11.12 The Differentiation of Blood Cells**
One type of adult stem cell gives rise to various forms of blood cells. These stem cells are found in the red bone marrow, where they divide. Some of the stem cells differentiate and change their gene expression to become a specific cell type. The differentiated blood cells are shown across the bottom of the image.

## 11.5 Biotechnology Ethics

Scientific advances frequently present society with ethical questions that must be resolved. For example, when first introduced, immunization and in vitro fertilization were highly controversial procedures. How will new technology be used safely? Who will benefit? Should the technology be used to make a profit? Biotechnology is no different.

Many feel that biotechnology is dangerous. There are concerns about contaminating the environment with organisms that are modified genetically in the lab. What would be the impact of such contamination? Biotechnology also allows scientists to examine molecularly the genetic characteristics of an individual. How will this ability to characterize individuals be used? How will it be misused? Others feel that biotechnology is akin to playing God.

### What Are the Consequences?

One way to explore the ethics of biotechnology is to weigh its pros against its cons. This method of thinking considers all the consequences and implications of biotechnology. Which outweighs the other? The benefits of nearly everything discussed in this chapter include a greater potential for better medical treatment, cures for disease, and a better understanding of the world around us. What price must we pay for these advances?

- The development of these technologies may mean that our personal genetic information becomes public record. How might this information be misused? Insurance companies might deny coverage or charge exorbitant premiums for individuals with genetic diseases.

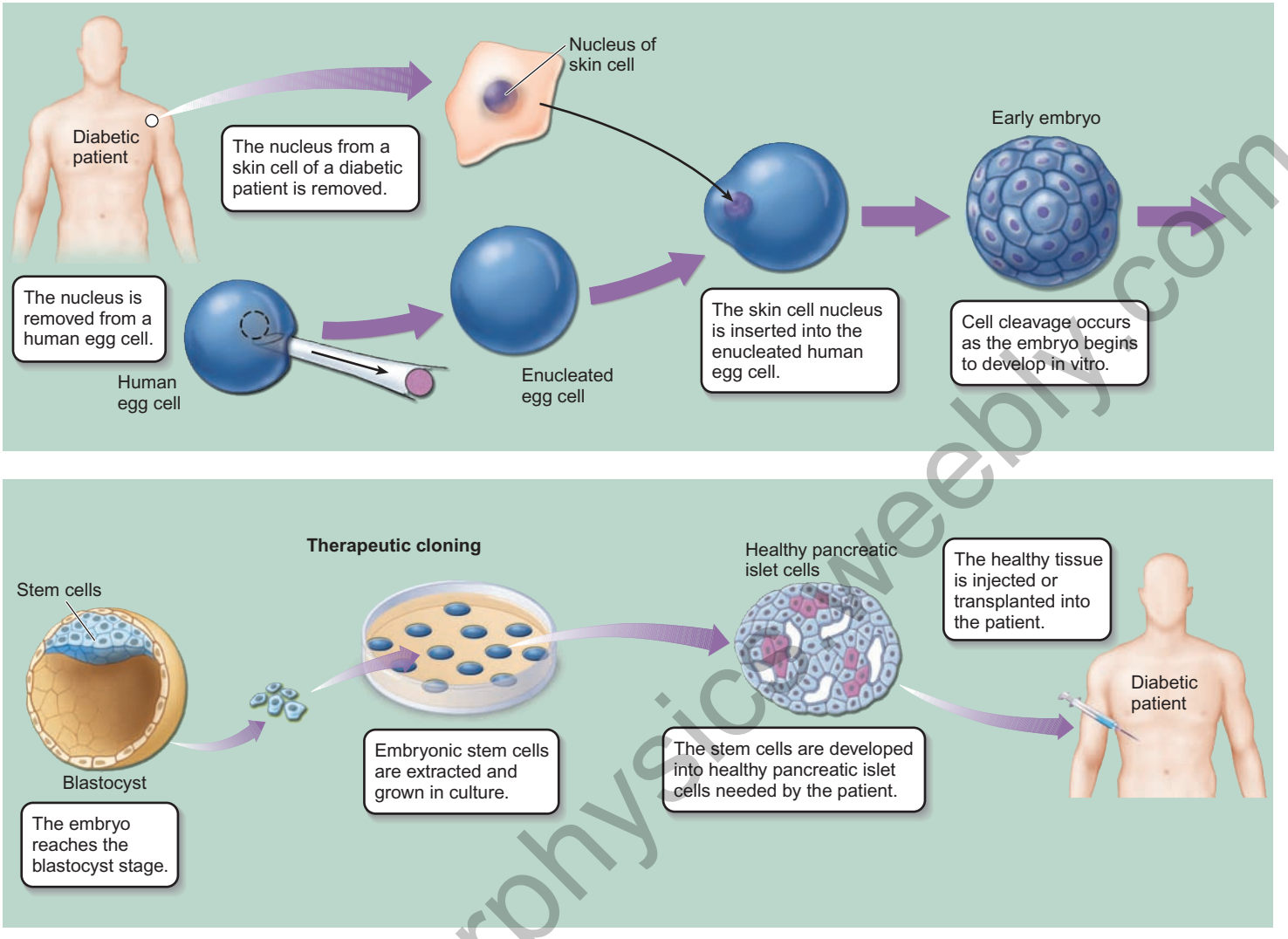



**FIGURE 11.13 Customized Stem Cell Lines**
One potential use of biotechnology is the production of customized stem cell lines. In this application a somatic cell from a patient would be inserted into a human egg from which the nucleus has been removed. The egg would divide and generate stem cells. These cells could then be cultured and used for therapy. In this example the stem cells could be used to create pancreatic cells to treat a diabetic patient.

- Cloning technology allows the creation of genetically modified foods that increase production and are more nutritious. Is this ethical if the genetically modified organism suffers because of disorders and pain caused by the change? Are you willing to risk the potential problems of a genetically modified organism becoming part of the ecosystem? How might the introduction of genetically modified species alter ecosystems and their delicate balance?

## Is Biotechnology Inherently Wrong?

Another way to explore the ethics of biotechnology is to ask if it violates principles that are valued by society. What aspects of biotechnology threaten the principles of the Bill of Rights? Basic human rights? Religious beliefs? Quality of life issues? Animal rights issues? Which of these sets of principles should be used to help us decide if biotechnology is ethical?

- Is it inherently wrong to produce genetically modified foods? Should companies be allowed to grow genetically modified crops? Should the foods be labeled as genetically modified when sold? How does this impact you as a consumer or simply as a person?
- Is it inherently wrong to manipulate genes? Are humans wise enough to use biotechnology safely? Is this something that only God should control? Would you have your child genetically altered as a fetus to prevent a genetic disease? Would you have your child genetically altered as a fetus to enhance desirable characteristics, such as intelligence, or even to control gender? Do you feel that one situation is morally justified but the other is not?
- Is it inherently wrong to harvest embryonic stem cells? Stem cells may provide new avenues of treatment for many disorders. Although there are several sources of stem cells, the cells of most interest are embryonic stem cells. Harvesting

these cells destroys the embryo. Even if the embryo is not yet aware of its environment and does not sense pain, is it ethical to use human embryos to advance the treatment of disease?
- Are we morally obligated to search for cures and treatments? Can we stop research if people still need treatment and cures?

Clearly, these are issues that our society will debate for some time. Many of these issues have been debated for decades and bring forward very strong feelings and very different world views. As you continue to hear more about biotechnology in your day-to-day life, consider how that form of biotechnology may affect you.

## 11.5 CONCEPT REVIEW

15. Match each of the following questions to the appropriate statements.

| Ethical Principle | Statements |
|---|---|
| What are the consequences? | The benefits of biotechnology more than compensate for its problems. |
| Is biotechnology inherently wrong? | Regardless of the benefits of biotechnology, we should not tamper with organisms in this way. |
| Is the manipulation of an organism's genes playing God? | Religion and science do not conflict with one another. |

16. List three of the benefits of biotechnology in your life today.

## Summary

Advances in biotechnology are possible because organisms use a common genetic language to make proteins. New techniques, such as DNA fingerprinting and DNA sequencing, allow scientists to compare DNA directly. These techniques involve multiple steps, including the polymerase chain reaction, the use of restriction enzymes, and electrophoresis. One large-scale analysis was the Human Genome Project. Scientists are hopeful that the information gained from the Human Genome Project will allow the better diagnosis and treatment of many medical conditions. The genomes of many other organisms have also been characterized, resulting in the new fields of biology called genomics, transcriptomics, and proteomics. The commonality of the genetic code allows DNA from one organism to be used by a different species. The techniques used to clone a gene and to clone an entire organism differ. Cloning a gene involves a number of techniques, including screening a DNA library. Cloning an organism involves somatic cell nuclear transfer. Stem cells have the potential to become multiple cell types. Many feel that the controlled growth of stem cells can be a medical treatment for many incurable medical conditions. The social concern surrounding biotechnology has created an ethical debate, which asks two fundamental questions:

- Do the benefits of biotechnology outweigh the problems?
- Are some aspects of biotechnology inherently wrong?

## Key Terms

*Use the interactive flash cards on the **Concepts in Biology**, 14/e website to help you learn the meaning of these terms.*

bioremediation 239
biotechnology 226
clone 235
DNA fingerprinting 226
DNA library 236
electrophoresis 227
gene therapy 239
genetically modified (GM) 235
genomics 235
multigene families 234
plasmid 236
polymerase chain reaction (PCR) 226
proteomics 235
recombinant DNA 235
restriction enzymes 227
restriction fragments 227
restriction sites 227
segmental duplications 234
somatic cell nuclear transfer 239
stem cells 240
tandem clusters 234
transcriptomics 235
transformation 235
variable number tandem repeats (VNTRs) 226
vector 236

## Basic Review

1. Information in DNA can code for the same protein in any organism. (T/F)
2. DNA fingerprinting
   a. directly examines nucleotide sequence.
   b. examines segments of DNA, which vary in length between individuals.
   c. transfers DNA from one person to another.
   d. uses stem cells.
3. Restriction fragments
   a. are used in a technique that sequences DNA.
   b. create many copies of DNA from a small amount.
   c. are pieces of DNA cut by enzymes at specific sites.
   d. are pieces of protein cut by enzymes at specific sites.

4. A technique that separates DNA fragments of different lengths is
   a. electrophoresis.
   b. DNA sequencing.
   c. polymerase chain reaction.
   d. DNA fingerprinting.
5. The Human Genome Project
   a. was an international effort.
   b. determined the sequence of a healthy human genome.
   c. allows comparisons of the human genome with that of other organisms.
   d. All of the above are correct.
6. The term *cloning* can be applied to which of the following situations?
   a. creating an exact copy of a fragment of DNA
   b. creating a second organism that is genetically identical to the first
   c. using a restriction enzyme
   d. Both a and b are correct.
7. Which of the following terms best describes an organism that possesses a cloned fragment of DNA from another species?
   a. uncloned
   b. genetically modified (GM)
   c. differentiated
   d. genomic
8. Stem cell research is controversial because
   a. of the source of stem cells.
   b. stem cells may cure certain diseases.
   c. stem cells are not yet differentiated.
   d. stem cells are not yet determined.
9. DNA libraries are
   a. stored in computers, so that they can be easily searched.
   b. are an index of various organisms.
   c. collections of DNA fragments that represent the genome of an organism.
   d. a person's unique electrophoresis banding pattern.
10. Restriction enzymes
    a. cut DNA randomly.
    b. cut DNA at specific sequences.
    c. can create sticky ends.
    d. Both b and c are correct.
11. _____ is the process a cell goes through to select which genes it will express.
12. This procedure removes a nucleus from a cell of the organism that will be cloned.
    a. somatic cell nuclear transfer
    b. transposition
    c. cloning
    d. electrophoresis
13. Scientists have used some types of _____ to transfer genes from one cell type to another.
14. _____ DNA is DNA that has been constructured by inserting new pieces of DNA into it from another organism.
15. This field of study examines the proteins that are predicted from the DNA sequence.
    a. genomics
    b. proteomics
    c. transcriptomics
    d. restriction enzyme technology

**Answers**
1. T 2. b 3. c 4. a 5. d 6. d 7. b 8. a 9. c 10. d 11. Determination 12. a 13. viruses or plasmids 14. Recombinant 15. b

## Thinking Critically

**Crime Scene Work with DNA**
An 18-year-old college student reported that she had been raped by someone she identified as a "large, tanned white man." A student in her biology class fitting that description was said by eyewitnesses to have been, without a doubt, in the area at approximately the time of the crime. The suspect was apprehended and, on investigation, was found to look very much like someone who lived in the area and who had a previous record of criminal sexual assaults.

Samples of semen from the woman's vagina were taken during a physical exam after the rape. Cells were also taken from the suspect.

He was brought to trial but was found to be innocent of the crime based on evidence from the criminal investigations laboratory. His alibi—that he had been working alone on a research project in the biology lab—held up. Without PCR genetic fingerprinting, the suspect would surely have been wrongly convicted, based solely on circumstantial evidence provided by the victim and the eyewitnesses.

Place yourself in the position of the expert witness from the criminal laboratory who performed the PCR genetic fingerprinting tests on the two specimens. The prosecuting attorney has just asked you to explain to the jury what led you to the conclusion that the suspect could not have been responsible for this crime. Remember, you must explain this to a jury of 12 men and women who, in all likelihood, have little or no background in the biological sciences.

CHAPTER 12

# Diversity Within Species and Population Genetics



## Conservation—Right Down To Your Genes

*Nature saves the best and to be shared by many.*

## CHAPTER OUTLINE



It has been estimated that there are 10,000,000,000 different kinds of genes distributed among Earth's living organisms. However, some of these genes are responsible for controlling similar biochemical pathways among many types of organisms. Biologists say these genes are "conserved." That is, they are similar and generally show little variation. For example, genes that control fundamental biochemical processes such as cellular respiration are strongly conserved across different kinds of organisms. The aerobic cellular respiration process carried out by the bacterium *Escherichia coli* is almost the same as that performed by human beings, *Homo sapiens*, and the maple tree, *Acer saccharum*. Other genes are more specialized and unique to certain species. For example, a certain strain of cholera bacteria (*Vibrio cholerae*) contains a unique gene allowing these bacteria to better survive as human pathogens. These genes have not been conserved across species.

Species that are very common, for example, *E. coli*, usually have great genetic diversity. Genetic diversity within the species makes it highly likely that the species will be able to adapt and survive in ever-changing environments. Species with a great deal of genetic diversity are also more likely to continue to exist for longer periods. On the other hand, species with limited diversity do not have the same genetic resources to cope with a changing environment, and are threatened with extinction. Many organisms on the verge of extinction, for example, cheetahs (*Acinonyx jubatus*), are in this position. In the past, cheetahs were known to be in Asia, Europe, and Africa. However, they now exist only in a small population in sub-Saharan Africa and in an even smaller group in northern Iran.

- **What is the value of genetic diversity?**
- **Why do some species have little genetic diversity?**
- **Are extinctions that humans cause a problem?**

## Background Check

**Concepts you should already know to get the most out of this chapter:**

- The molecular basis of heredity (chapter 8)
- The source of genetic diversity (chapter 9)
- How meiosis, genes, and alleles are related to one another (chapter 10)
- Mendel's laws of inheritance (chapter 10)

# 12.1 Genetics in Species and Populations

Plants, animals, and other kinds of organisms exist not only as genetic individuals but also as part of a larger, interbreeding group. An understanding of two terms—*species* and *population*—is necessary, because these are interconnected. A **species** consists of all the organisms potentially capable of breeding naturally among themselves and having offspring that also interbreed successfully. As of 2009, the results of a world-wide effort (Catalogue of Life) to record all species has identified over 1,160,000 species. A **population** is a group of organisms that are potentially capable of breeding naturally and are found in a specified area at the same time.

The concept of a species accounts for individuals from *different* populations that interbreed successfully. Since biologists designate populations as the organisms found in a particular place at a particular time, most populations consist of only a portion of all the members of the species. For example, the dandelion population in a city park on the third Sunday in July is only a small portion of all dandelions on the planet. However, a population can also be all the members of a species—for example, the human population of the world in 2008 or all the current members of the endangered whooping cranes.

**Population genetics** is the study of the kinds of genes within a population, their relative numbers, and how these numbers change over time. This information is used as the basis for classifying organisms and studying evolutionary change. From the standpoint of genetics, a population consists of a large number of individuals, each with its own set of alleles. However, the populations may contain many more different alleles than any one member of the species. Any one organism has a specific genotype consisting of all the genetic information that organism has in its DNA. A diploid organism has a maximum of 2 different alleles for a gene, because it has inherited an allele from each parent. In a population, however, there may be many more than 2 alleles for a specific characteristic. In humans, there are 3 alleles for blood type (A, B, and O) within the population, but an individual can have only up to 2 of the alleles (figure 12.1).

Theoretically, all members of a population are able to exchange genetic material. Therefore, we can think of all the genetic information of all the individuals of the same group as a *gene pool*. A **gene pool** consists of all the alleles of all the individuals in a population. Because each organism is like a container of a particular set of these alleles, the gene pool contains much more genetic variation than does any one of the individuals. A gene pool is like a gumball machine containing red, blue, yellow, and green balls (alleles). For 25 cents and a turn of the knob, two gumballs are dispensed from the machine. Two red gumballs, a red and a blue, a yellow and a green, or any of the other possible color combination may result from any one gumball purchase. A person buying gumballs will receive no more than 2 of the 4 possible gumball colors and only 1 of 10 possible color combinations. Similarly, individuals can have no more than 2 of the many alleles for a given gene contained within the gene pool and only 1 of several possible combinations of alleles.


**FIGURE 12.1 Genetic Diversity in Individuals and Populations**
Any individual can have only 2 alleles for a particular gene, but the population can have several alleles of that gene.

Gene Pools

## 12.1 CONCEPT REVIEW

1. How do the concepts of *species* and *genetically distinct populations* differ?
2. Why is it that a population can be more genetically diverse that an individual?
3. Give an example of a gene pool containing a number of separate populations.

## 12.2 The Biological Species Concept

A species is a population of organisms that share a gene pool and are reproductively isolated from other populations. This definition of a species is often called the **biological species concept**; it involves the understanding that organisms of different species do not interchange genetic information—that is, they don't reproduce with one another. *An individual organism is not a species but, rather, is a member of a species;* some people refer to the "male" or "female" species. This is an incorrect understanding of the species concept. A correct statement would be, "the male of the species." A clear understanding of the concept of species is important as we begin to consider how genetic material is passed around within populations as sexual reproduction takes place. It will also help in considering how evolution takes place.

If we examine the chromosomes of reproducing organisms, we find that they are equivalent in number and size and usually carry very similar groups of genes. In the final analysis, the biological species concept assumes that the genetic similarity of organisms is the best way to identify a species, regardless of where or when they exist. Individuals of a species usually are not evenly distributed within a geographic region but, rather, occur in clusters as a result of barriers that restrict movement or the local availability of resources. Local populations with distinct genetic combinations may differ quite a bit from one place to another. There may be differences in the kinds of alleles and the numbers of each kind of allele in different populations of the same species. Note in figure 12.2

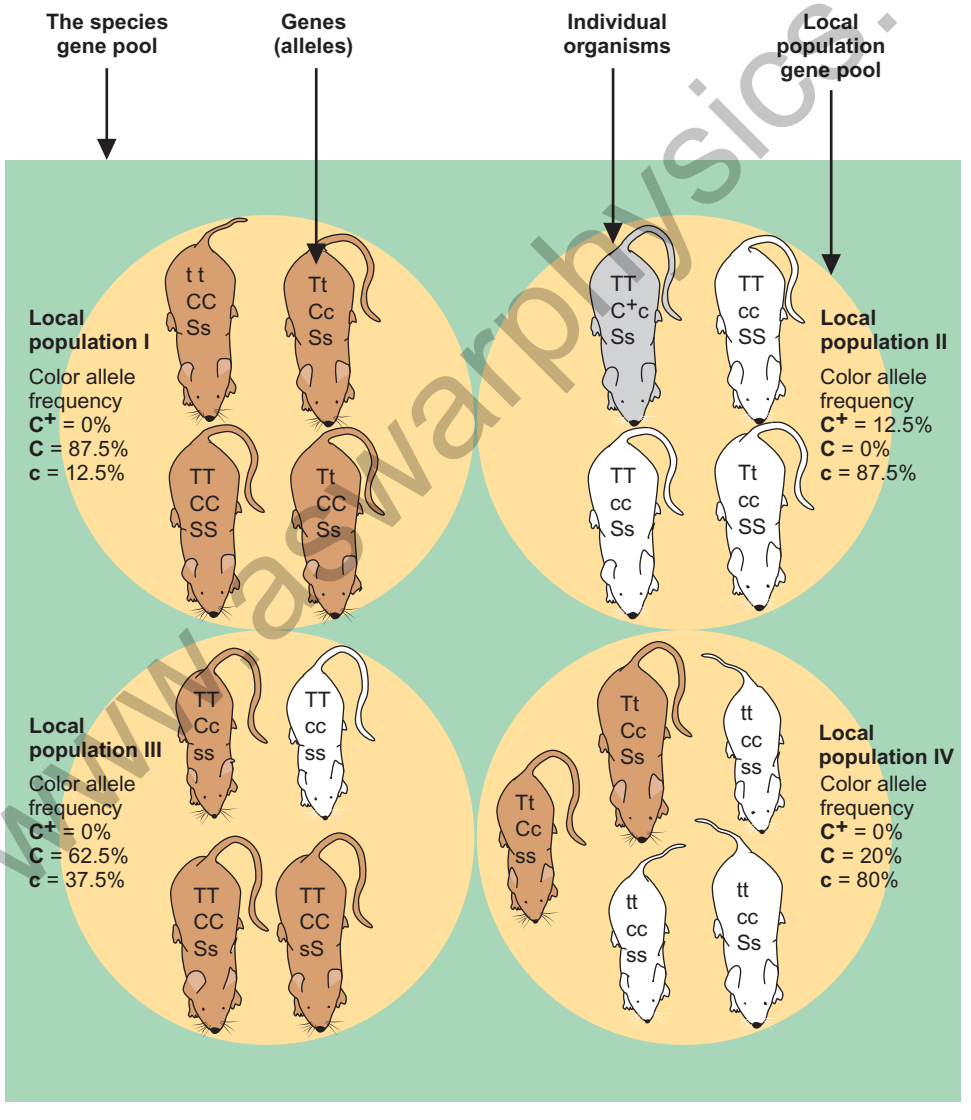



**FIGURE 12.2 Genes, Populations, and Gene Pools**
Each individual shown here has a specific combination of alleles that constitutes its genotype. The frequency of a specific allele varies from one local population to another. Each local population has a gene pool that is somewhat different from the others. Notice how differences in the frequencies of particular alleles in local populations affect the appearance of the individuals. *Assume that* $T$ = long tail; $t$ = short tail; $C^+$ = gray color; $C$ = brown color; $c$ = white color; $S$ = large size; and $s$ = small size.

that, within the gene pool of the species, there are 3 possible alleles for color ($C^+$, $C$, and $c$). However, how often these alleles appear in the population—the frequencies of these alleles—is different in the four local populations, and the difference in how often an allele occurs is reflected in the colors seen in the individuals of the population.

## Gene and Allele Frequencies

**Gene frequency** and **allele frequency** are defined as how often an allele is found in a population. In general, the term *gene frequency* is used when describing the idea that there are genetic differences between populations. The term *allele frequency* is used when specifically discussing how common a particular form of a gene (allele) is compared, with other forms. For example, the frequency of the blond hair allele is high in northern Europe but low in Africa.

Allele frequency is commonly stated in terms of a percentage or a decimal fraction (e.g., 10%, or 0.1; 50%, or 0.5). It is a mathematical statement of how frequently an allele is found in a population. It is possible for two populations of the same species to have all the same alleles, but with very different frequencies.

As an example, all humans are of the same species and, therefore, constitute one, large gene pool found on Earth. There are, however, many distinct, local populations scattered around the world. These, more localized populations show many distinguishing characteristics, which have been perpetuated from generation to generation. In Africa, alleles for dark skin, tightly curled hair, and a flat nose have very high frequencies. In Europe, the allele frequencies for light skin, straight hair, and a narrow nose are the highest (Outlooks 12.1). People in Asia tend to have moderately colored skin, straight hair, and broad noses (figure 12.3). All three of these populations have alleles for dark skin and light skin, straight hair and curly hair, narrow noses and broad noses. The three differ, however, in the frequencies of these alleles. Once a mixture of alleles is present in a population, that mixture tends to maintain itself, unless something changes the frequencies. In other words, allele frequencies do not change without reason. With the development of transportation, more people have moved from one geographic area to another, and human allele frequencies have begun to change. Ultimately, as barriers to interracial marriage (both geographic and sociological) are leveled, the human gene pool will show fewer and fewer geographically distinct populations.

People think that allele frequency has something to do with dominance and recessiveness, but this is not true. Often in a population, recessive alleles are more frequent than their dominant counterparts. Straight hair, blue eyes, and light skin are all recessive characteristics, yet they are quite common in the populations of certain European countries. See table 12.1 for other examples. What really determines the frequency of an

**TABLE 12.1 Recessive Traits with a High Frequency of Expression**

Many recessive characteristics are extremely common in some human populations. The corresponding dominant characteristic is also shown here.

| Recessive | Dominant |
|---|---|
| Light skin color | Dark skin color |
| Straight hair | Curly hair |
| Five fingers | Six fingers |
| Type O blood | Type A or B blood |
| Normal hip joints | Dislocated hip birth defect |
| Blue eyes | Brown eyes |
| Normal eyelids | Drooping eyelids |
| No tumor of the retina | Tumor of the retina |
| Normal fingers | Short fingers |
| Normal thumb | Extra joint in the thumb |
| Normal fingers | Webbed fingers |
| Ability to smell | Inability to smell |
| Normal tooth number | Extra teeth |
| Presence of molars | Absence of molars |


**FIGURE 12.3 Allele Frequency Differences Among Humans**
Different physical characteristics displayed by people from different parts of the world are an indication that allele frequencies differ as well.

## OUTLOOKS 12.1

### Your Skin Color, Gene Frequency Changes, and Natural Selection

For centuries we have classified humans into "races" based on superficial traits. One of the most obvious is skin color. Fill out any survey and you will probably find a category asking you to identify yourself according to race. Skin color is almost always what comes to mind when you decide if you are Caucasian, African-American, Hispanic, or mixed. But are there genes involved in this trait? When did humans develop different skin colors? What factors may have led to and stabilized the existence of different-colored groups of people?

Yes, skin color is regulated by your genes. In fact, several genes are involved in the polygenic inheritance of pigment production. Scientists had thought that beginning about 40,000 years ago, modern humans in Europe began to grow paler as they migrated farther north. They hypothesized that pale skin allows more sunlight to penetrate the skin. This allowed more UV light to stimulate the production of vitamin D, used in bone growth and many other essential pathways. One of the genes that apparently causes pale skin, known as SLC24A5, has been identified in many Europeans (but not in Asians). There are two forms of this gene that control the production of proteins used for skin pigmentation. The proteins produced by these two alleles differ by only one amino acid. Almost all Africans and East Asians carry the "dark" form of the gene, whereas 98% of Europeans have the other—the "pale" gene. Careful analysis of DNA now suggests that the source of the pale gene was a mutation and that it increased in frequency in European populations more recently than previously thought, most likely between 6,000 and 12,000 years ago.

allele in a population is the allele's value to the organisms possessing it. Dark-skin alleles are valuable to people living under the bright sun in tropical regions. These alleles are less valuable to those living in the less intense sunlight of the cooler European countries. This idea of the value of alleles and how it affects allele frequency will be dealt with more fully when the process of natural selection is discussed in chapter 13.

## Subspecies, Breeds, Varieties, Strains, and Races

Within a population, genetic material is repackaged into new individuals from one generation to the next. Often, there is very little adding or subtracting of genetic material from a local population of organisms, and a widely distributed species consists of a number of more or less separate groups, known as **subspecies** (or **breeds, varieties, strains,** or **races**). All of these terms are used to describe distinct populations within a species. However, certain terms are used more frequently than others, depending on one's field of interest. For example, dog breeders use the term *breed*, horticulturalists use the term *variety*, microbiologists use the term *strain*, and anthropologists use the term *race* (Outlooks 12.2). The most general and most widely accepted term is *subspecies*. Look again at figure 12.2. The gene pool of the species consists of all the alleles of all individuals of the 4 separate populations. A local that shows differences from other local populations is considered a subspecies. For example, the American robin is found throughout North America from southern Canada to central Mexico (figure 12.4). Of the seven subspecies of this common thrush (*Turdus* = *thrush*),

(a) ***Turdus migratorius***

(b) ***Turdus migratorius confirmis***

**FIGURE 12.4 Subspecies of the American Robin (*Turdus migratorius*).** *(a)* While six of the subspecies of this thrush have the distinctive red breast, the San Lucas subspecies *(b)*, located in the highlands of the Cape District of Southern Baja California, Mexico, has an under belly that is a pale gray-brown.

## OUTLOOKS 12.2

### Biology, Race, and Racism

The concept of racial difference among groups of people must be approached carefully. Three distortions can occur when people use the term *race*. First, the designation of race focuses on differences, most of which are superficial. Skin color, facial features, and hair texture are examples. Although these characteristics are easy to see, they are arbitrary, and an emphasis on them tends to obscure the fact that humans are all fundamentally the same, with minor variations in the frequency of certain alleles. A second problem with the concept of race is that it is very difficult to separate genetic from cultural differences among people. People tend to equate cultural characteristics with genetic differences. Culture is learned and, therefore, is an acquired characteristic not based on the genes a person inherits. Cultures do differ, but these differences cannot be inherited and therefore used as a basis for claiming genetic distinctions. Third, a study of the human genome has revealed that there are usually more genetic differences within so-called racial groups than between them. Because of such distortions, the concept that humans can be divided into racial groups is no longer popular among scientists.

**Changing attitudes**

the San Lucas robin, *Turdus migratorius confinis* is found in the highlands of Baja California. It is distinct from the others in having a pale gray-brown under belly instead of the 'robin red breast.'

Most robins have alleles for red breast coloration; very few individuals have alleles for gray-brown. Since the San Lucas robins are geographically isolated from the main gene pool, they only mate with one another. Thus, the different color patterns result from a higher incidence of gray-brown color alleles in the Baja California populations and a high frequency of the red breast alleles in the other populations.

### 12.2 CONCEPT REVIEW

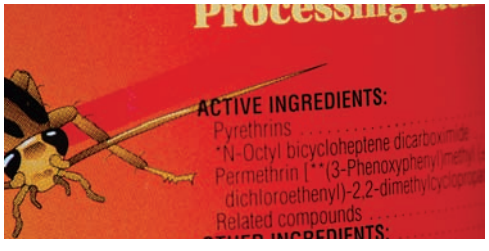
4. Describe the biological species concept.
5. What is meant by the terms *gene frequency* and *allele frequency*?
6. How are the terms *gene* and *allele frequency* used differently?
7. Give an example of a human characteristic that has a high frequency in Europe and a low frequency in Africa.

## 12.3 How Genetic Diversity Comes About

**Genetic diversity** is a term used to describe genetic differences among members of a population. *High genetic diversity* indicates many different kinds of alleles for each characteristic, and *low genetic diversity* indicates that nearly all the individuals in the population have the same alleles. A large gene pool with high genetic diversity is more likely to contain some genetic combinations that will allow the organisms to adapt to a new environment; whereas low genetic diversity can have devastating consequences. A number of mechanisms introduce genetic diversity into a population.

### Mutations

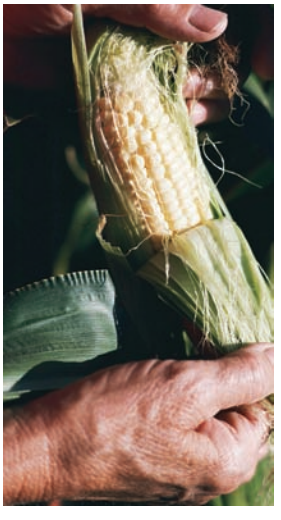
*Mutations* introduce new genetic information into a population by modifying alleles that are already present. Sometimes, a mutation introduces a new allele into the gene pool of a species. At other times, a mutation may introduce an allele that was absent in a local population, although it is present in other populations of the species. All the different alleles for a trait originated as a result of mutations some time in the past and have been maintained within the gene pool of the species as they have been passed from generation to generation during reproduction. Many mutations are harmful, but very rarely one will occur that is valuable to the organism. If a mutation produces a harmful allele, the allele remains uncommon in the population. For example, the *Anopheles* mosquito is responsible for transmitting malaria in many African countries. At some point in the past, mutations occurred in the DNA of these mosquitoes that



made some individuals tolerant to the insecticide Pyrethrin, even before the chemical had been used. These alleles remained very rare in these insect populations until Pyrethrin was used. Then, these alleles became very valuable to the mosquitoes that carried them. Because the mosquitoes that lacked the alleles for tolerance died when they came into contact with Pyrethrin, more of the Pyrethrin-tolerant individuals were left to reproduce the species; therefore, the Pyrethrin-tolerant alleles became much more common in these populations. Scientists have recently found up to 90% Pyrethrin resistance in *Anopheles* mosquitoes that live in several African countries.

## Sexual Reproduction

Although the process of *sexual reproduction* does not create new alleles, it tends to generate new *genetic combinations* when the genetic information from two individuals mixes during fertilization, generating a unique individual. This doesn't directly change the frequency of alleles within the gene pool. However, the new member may have a unique combination of characteristics. This combination may be so superior to those of other members of the population that the new member will be much more successful in producing offspring. In a corn population, for example, there may be alleles for resistance to corn blight (a fungal disease) and to attack by insects. Corn plants that possess both of these characteristics will be more successful than corn plants that have only one of these qualities. They will probably produce more offspring (corn seeds) than the others, because they will survive both fungal and insect attacks. Thus, there will be a change in the allele frequency for these characteristics in future generations.



Corn

## Migration

The *migration* of individuals from one genetically distinct population to another is also an important way for alleles to be added to or subtracted from a local population. Whenever an organism leaves one population and enters another, it subtracts its genetic information from the population it left and adds it to the population it joins. If it contains rare alleles, it may significantly affect the allele frequency of both populations. The extent of migration need not be great; however, as long as alleles are entering or leaving a population, the gene pool will change.

Many animal populations in zoos are in danger of dying out because of severe inbreeding or line breeding (breeding with near relatives), resulting in reduced genetic diversity (figure 12.5). Often, when genetic diversity is reduced, deleterious recessive alleles in closely related mates are passed to offspring in a homozygous state, resulting in offspring that have reduced chances of survival. Most zoo managers have recognized the importance of increasing genetic diversity in their small populations of animals and have instituted programs of loaning breeding animals to distant zoos in an effort to increase genetic diversity. In effect, they are attempting to simulate the natural migration that frequently introduces new alleles from distant populations. Captive breeding program have been critical in saving many species, including Guam rails, California condors, Przewalski's horses, scimitar-horned oryx, *Partula* snails, and Spix's macaws.

**FIGURE 12.5 Captive Breeding of the Black-Footed Ferret**
In October 1985, the Wyoming Game and Fish Department, in cooperation with the U.S. Fish & Wildlife Service, started the captive breeding program for North America's most endangered mammal, the black-footed ferret. Attention was paid to making sure that as much genetic variation was retained as possible—for example, they maintained a sperm bank of particularly valuable males. As a result, the successful return of black-footed ferrets to the plains of the American West began in 1991. The total wild population of black-footed ferrets in 2007 was over 750 individuals in the United States. Biologists hope that a new population census will show a further increase in wild black-footed ferrets. However, biologists still fear that a lack of genetic diversity may jeopardize the populations.

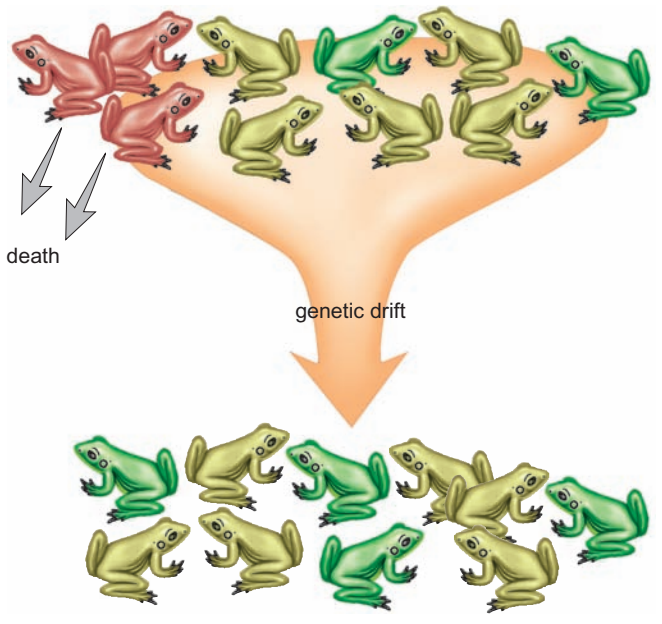



**FIGURE 12.6 Genetic Drift**
The gene pool of a small population may not have the same proportion of alleles as the previous generation. Notice that in the original population, the red frogs were eliminated and failed to breed. Therefore, their genes were not passed on to the next generation. As a result, the frequencies of the genes change in the gene pool.

## The Importance of Population Size

The *size of the population* has much to do with how effective any mechanism is at generating diversity within a gene pool. The smaller the population, the less genetic diversity it can contain. Therefore, migrations, mutations, and accidental death can have great effects on the genetic makeup of a small population. For example, if a town has a population of 20 people and only 2 have brown eyes and the rest have blue eyes, what happens to those 2 brown-eyed people is more critical than if the town has 20,000 people and 2,000 have brown eyes. Although the ratio of brown eyes to blue eyes is the same in both cases, even a small change in a population of 20 could significantly change the frequency of the brown-eye allele. Often, in small populations, random events can significantly alter the gene pool when rare alleles are lost from the population. This process is called **genetic drift** because the changes are not caused by selection (figure 12.6). This idea will be discussed in greater detail in chapter 13.

### 12.3 CONCEPT REVIEW

8. Why can there be greater genetic diversity within a gene pool than in an individual organism?
9. List three mechanisms that contribute to genetic diversity.

# 12.4 Why Genetically Distinct Populations Exist

Many species have wide geographic distribution with reasonably distinct subspecies. There are four reasons that these subspecies developed: adaptation to local environmental conditions, the founder effect, a genetic bottleneck, and barriers to movement.

## Adaptation to Local Environmental Conditions

Because organisms within a population are not genetically identical, some individuals may possess genetic combinations that are valuable for survival in the local environment. As a result, some individuals find the environment less hostile than do others. The individuals with unfavorable genetic combinations leave the population more often, either by death or migration, and remove their genes from the population. Therefore, local populations that occupy sites that differ greatly from conditions at other locations would be expected to consist of individuals having gene combinations suited to local conditions. For example, White Sands National Monument in New Mexico has extensive dunes of white gypsum sand. Several of the animals that live there, such as lizards and mice, have very light coloring, which allows them to blend in with their surroundings. Other populations of the same species that do not live in such a white environment do not have the light coloring.

Many kinds of animals that live in caves lack pigments and eyes, including salamanders, flatworms, shrimp, fish, beetles, and crayfish. A blind Texas salamander (*Eurycea rathbuni*) is at a severe disadvantage when venturing above ground. A blind Texas salamander living in a cave where there is no light, however, is not at the same disadvantage. Thus, these two environments might allow or encourage characteristics to be present in the two populations at different frequencies. Because it takes energy to produce eyes and pigment, individual cave-dwelling Texas blind salamanders that do not produce these characteristics may have an advantage over the individuals that continue to spend the energy to produce these features. (figure 12.7).

## The Founder Effect

The second mechanism that creates genetically distinct populations with unique allele frequencies is the founding of a new population and, so, is called the *founder effect*. The **founder effect** is a form of genetic drift, in which a genetically distinct local population is established by a few colonizing individuals carrying with them alleles that differ in frequencies from those in their original population. The collection of alleles of a small founding population is likely to be different from that present in the larger parent population from which it came. After all, a few pioneering individuals leaving a population would be unlikely to carry copies of all the alleles found within the original population.

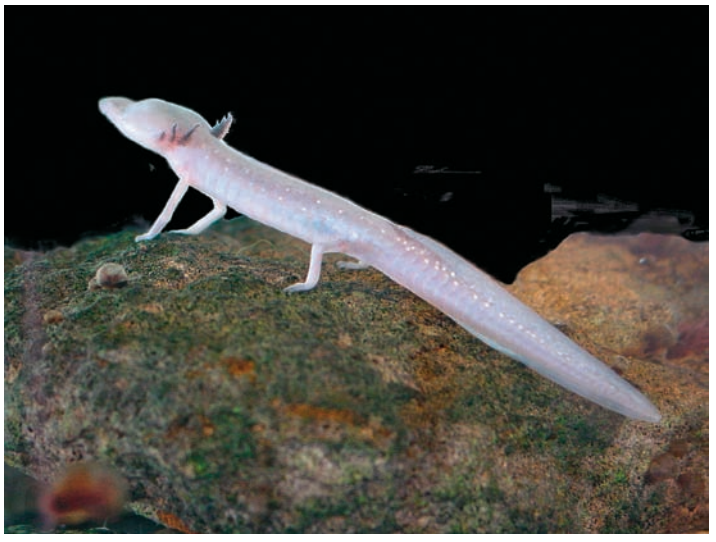


**FIGURE 12.7 Specialized Local Populations**
Many populations of animals that live in caves where there is no light lack pigment and functional eyes. If, in the past, the genetic material that directed the production of these traits was lost or mutated, it would not have negatively affected the organism. In fact, it may have been an advantage to not spend the energy to produce such unneeded traits. Hence, in many cave populations, there is a low frequency of genes for the production of eyes and pigment. This is a Texas blind salamander, an endangered species found only in the Edwards Aquifer underneath and near the city of San Marcos, Texas.

They may even carry an unrepresentative mixture of alleles. For example, the water snake *Nerodia sepidon* is found throughout the eastern part of the United States and extends into Canada. The northern water snake subspecies is widespread in the north-central United States and adjacent Canada and is generally brown with light diamond-shaped patches. The Lake Erie water snake subspecies is limited to the islands in the western section of Lake Erie. It is generally a solid color without the lighter patches. The difference in color patterns is related to different allele frequencies for color pattern. The Lake Erie water snake may have been founded by a small number of individuals from the mainland that had a high frequency of alleles for solid coloration rather than the more typical banded pattern. (It is even possible that the island populations could have been founded by one fertilized female.) Once a small founding population establishes itself, it tends to maintain its collection of alleles, because the organisms mate only among themselves. This results in a reshuffling of alleles from generation to generation but does not introduce new genetic information into the population.

## Genetic Bottleneck

The third cause of local, genetically distinct populations relates to the history of the population. A **genetic bottleneck** is a form of genetic drift, in which there is a sharp reduction in population size due to a chance event that results in a reduction in genetic diversity in subsequent generations. When the size of a population is greatly reduced, some alleles will probably be lost from the population. Any subsequent increase in the size of the population by reproduction among the remaining members of the population will not replace the lost genetic diversity. For example, in the late 1800s the whooping crane population was estimated to be 1300 birds (figure 12.8). Their numbers decreased because of hunting and industrial development. By 1941 there were only 22 wild whooping cranes left in North America. Today there are over 400 birds. However, their lack of genetic diversity puts them at risk of extinction. Thousands of other species are currently undergoing genetic bottlenecks.

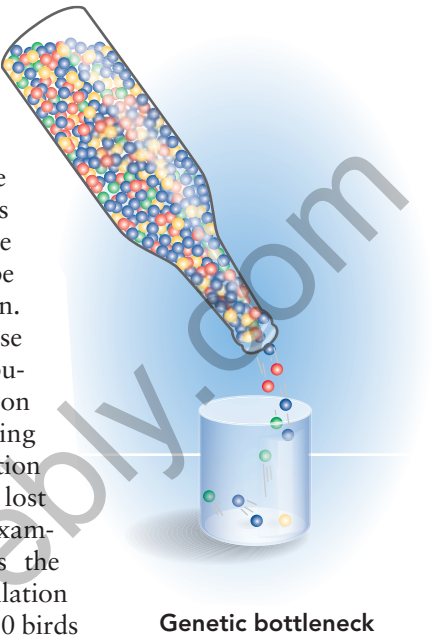



**FIGURE 12.8 Whooping Crane**
Today, the only remaining natural, self-sustaining flock of whooping cranes is in Wood Buffalo National Park, Canada and winters in Aransas National Wildlife Refuge, Texas. This flock had only 16 birds during the winter of 1941–1942, and numbered under 35 birds for the next two decades.

Although some endangered species were always rare, most have experienced recent reductions in their populations and a reduction in their genetic diversity, which is a consequence of severely reduced population size.

## Barriers to Movement

The fourth factor that tends to encourage the maintenance of genetically distinct populations is the presence of barriers to free movement. Animals and plants that live in lakes tend to be divided into small, separate populations by barriers of land. Whenever such barriers exist, there will very likely be differences in the allele frequencies from lake to lake, because each lake was colonized separately and the lakes' environments are not identical. Other species of organisms, such as migratory birds (e.g., Canadian geese, mallard ducks), experience few barriers; therefore, subspecies are not as common.

### 12.4 CONCEPT REVIEW

10. List four processes that can lead to local, genetically distinct populations.
11. In what way are the founder effect and a genetic bottleneck similar in their effect on the genetic diversity of a local population?

# 12.5 Genetic Diversity in Domesticated Plants and Animals

Humans often work with small, select populations of plants and animals in order to artificially construct specific genetic combinations that are useful or desirable. This is true of plants and animals used for food. If we can produce domesticated animals and plants with genetic characteristics for rapid growth, high reproductive capacity, resistance to disease, and other desirable characteristics, we can supply ourselves with energy in the form of food. Several processes are used to develop such specialized populations of plants and animals. Most have the side effect of reducing genetic diversity.

## Cloning

Recall that cloning is the process of reproducing organisms asexually, so that large numbers of genetically identical individuals are produced. These individuals are called *clones*.

Plants are easy to work with in this manner, because we can often increase the numbers of specific organisms by asexual (without sex) reproduction. Potatoes, apple trees, strawberries, and many other plants can be reproduced by simply cutting the original plant into a number of parts and allowing these parts to sprout roots, stems, and leaves. If a single potato has certain desirable characteristics, it can be reproduced asexually. All of the individual potato plants reproduced asexually would be genetically identical and would show the same desired characteristics. Figure 12.9 shows how a clone can be developed.

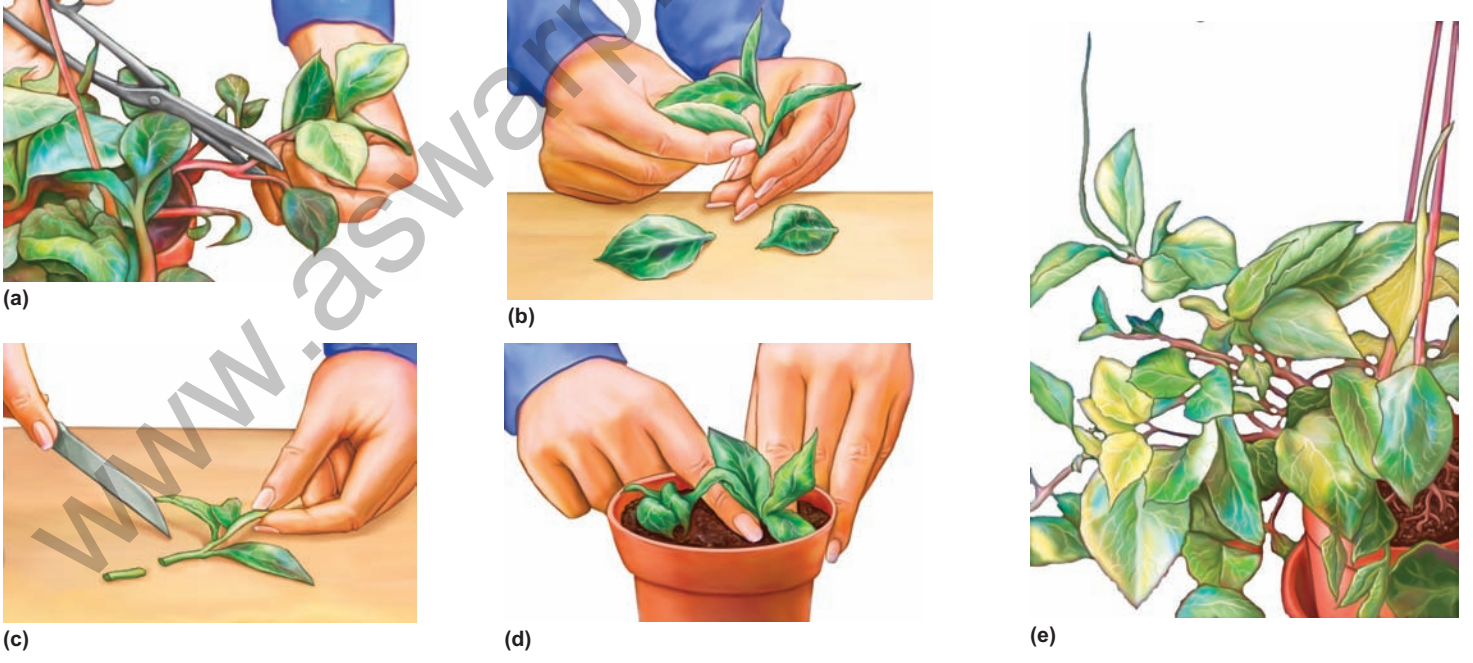



**FIGURE 12.9 Cloning Plants from Cuttings**
All the *Peperomia* houseplants in the last figure *(e)* were produced asexually from cuttings and are identical genetically. *(a–c)* The original plant is cut into pieces. Then, *(d)* the cut ends are treated with a growth stimulant and placed in moist soil. *(e)* Eventually, the pieces root and grow into independent plants.

**TABLE 12.2 Cloned Animals**

| Animal Cloned | Date First Cloned | Country Where Cloning Occurred |
|---|---|---|
| 1. Camel | 2009 | United Arab Emirates |
| 2. Carp | 1963 | China |
| 3. Cat | 2001 | United States |
| 4. Cattle | 2001 | United States |
| 5. Deer | 2003 | United States |
| 6. Dog | 2009 | South Korea |
| 7. Ferret | 2009 | United States |
| 8. Frog tadpole | 1962 | England |
| 9. Fruit fly | 2004 | Canada |
| 10. Gaur | 2001 | United States |
| 11. Goat | 2009 | Iran |
| 12. Horse | 2003 | Italy |
| 13. Mice | 1986 | Former U.S.S.R. |
| 14. Mouflon | 2001 | Italy |
| 15. Mule | 2003 | United States |
| 16. Pig | 2000 | Scotland |
| 17. Rabbit | 2003 | France |
| 18. Rat | 2003 | France |
| 19. Rhesus monkey | 2000 | United States |
| 20. Sheep | 1996 | Scotland |
| 21. Water buffalo | 2009 | China |
| 22. Wolf | 2009 | Korea |

The cloning of most kinds of domesticated animals is much more difficult than the cloning of plants. Even though this is not as yet a practical method of producing animals, in recent years, many kinds of animals have been cloned (table 12.2). The process involves the substitution of a nucleus from a mature animal for the nucleus of an egg. This cell is then stimulated to develop as an embryo. There is a high rate of failure, but the goals of animal cloning are the same as those of plant cloning: the production of genetically identical individuals.

## Selective Breeding

Humans can bring together specific genetic combinations in either plants or animals by selective breeding. Because sexual reproduction tends to generate new genetic combinations rather than preserve desirable combinations, the mating of individual organisms must be controlled to obtain the desirable combination of characteristics. *Selective breeding* involves the careful selection of individuals with specific desirable characteristics and their controlled mating, with the goal of producing a population that has a high proportion of individuals with the desired characteristics.

Through selective breeding, some varieties of chickens have been developed that grow rapidly and are good for meat. Others have been developed to produce large numbers of eggs. Often, the development of new varieties of domesticated animals and plants involves the crossing of individuals from different populations. For this technique to be effective, the desirable characteristics in each of the two varieties should have homozygous genotypes. In small, controlled populations, it is relatively easy to produce individuals that are homozygous for a specific trait. To make two characteristics homozygous in the same individual is more difficult. Therefore, such varieties are usually developed by crossing two different populations to collect several desirable characteristics in one organism. **Intraspecific hybrids** are organisms that are produced by the controlled breeding of separate varieties of the same species.

Occasionally, **interspecific hybrids**—hybrids between two species—are produced as a way of introducing desirable characteristics into a domesticated organism. Because plants can be reproduced by cloning, it is possible to produce an interspecific hybrid and then reproduce it by cloning. For instance, the tangelo is an interspecific hybrid between a tangerine and a grapefruit. An interspecific hybrid between cattle and the American bison was used to introduce certain desirable characteristics into cattle.

**Beefalo**

## Genetic Engineering

In recent years, scientific advances in understanding DNA have allowed specific pieces of genetic material to be inserted into cells. This has greatly expanded scientists' ability to modify the characteristics of domesticated plants and animals. The primary goal of genetic engineering is to manipulate particular pieces of DNA and transfer them into specific host organisms, so that they have certain valuable characteristics. These topics were dealt with in greater detail in chapter 11.

## The Impact of Monoculture

Although some of the previously mentioned techniques have been used to introduce new genetic information into domesticated organisms, one of the goals of domestication

**FIGURE 12.10 Monoculture**
This wheat field is an example of monoculture, a kind of agriculture in which large areas are exclusively planted with a single crop with a very specific genetic makeup. Monoculture makes it possible to use large farm machinery, but it also creates conditions that can encourage the spread of disease because the plants have reduced genetic diversity.

is to produce organisms that have uniform characteristics. In order to achieve such uniformity, it is necessary to reduce genetic diversity. Agricultural plants have been extremely specialized through selective breeding to produce the qualities that growers want. Most agriculture in the world is based on extensive plantings of the same varieties of a species over large expanses of land. This agricultural practice is called **monoculture** (figure 12.10). It is certainly easier to manage fields in which only one kind of plant is growing, especially when herbicides, insecticides, and fertilizers are tailored to meet the needs of specific crop species. However, with monoculture comes a significant risk. Because these organisms are so similar, if a new disease comes along, most of them will be affected in the same way and the whole population may be killed or severely damaged.

Our primary food plants and domesticated animal species are derived from wild ancestors that had genetic combinations that allowed them to compete successfully with other organisms in their environment. When humans reduce genetic diversity by developing special populations with certain desirable characteristics, other valuable genetic information is lost from the gene pool. When we select specific, good characteristics, we often get harmful ones along with them. Therefore, these "special" plants and animals require constant attention. Insecticides, herbicides, cultivation, and irrigation are all used to aid the plants and animals we need. In effect, these plants are able to live only under conditions that people carefully maintain (figure 12.11).

Because our domesticated organisms are so genetically similar, there is a great danger that an environmental change or new disease could cause great damage to our ability to produce food. In order to protect against such disasters, gene banks have been established. Gene banks consist of populations of primitive ancestors of modern domesticated plants and animals (figure 12.12). By preserving these organisms, their genetic diversity is available for introduction into our domesticated plants and animals if the need arises.

**FIGURE 12.11 Cash Crops Require Constant Attention**
The photograph shows a portion of a plantation where native forest has been cleared for the planting of bananas. However, without constant attention, native rainforest plants encroach back into the bananas.

## 12.5 CONCEPT REVIEW

12. How do the genetic combinations in clones and sexually reproducing populations differ?
13. How is a clone developed? What are its benefits and drawbacks?
14. How is an intraspecific hybrid formed? What are its benefits and drawbacks?
15. Why is genetic diversity in domesticated plants and animals reduced?

**FIGURE 12.12 The Banking of Genes**
Plant growers and breeders send genetic material, such as seeds, to the National Seed Storage Laboratory in Ft. Collins, Colorado, where it is stored at extremely cold temperatures to prevent deterioration. Gene banks will play an ever-increasing role in preserving biodiversity as the rate of extinction increases.

## 12.6 Is It a Species or Not? The Evidence

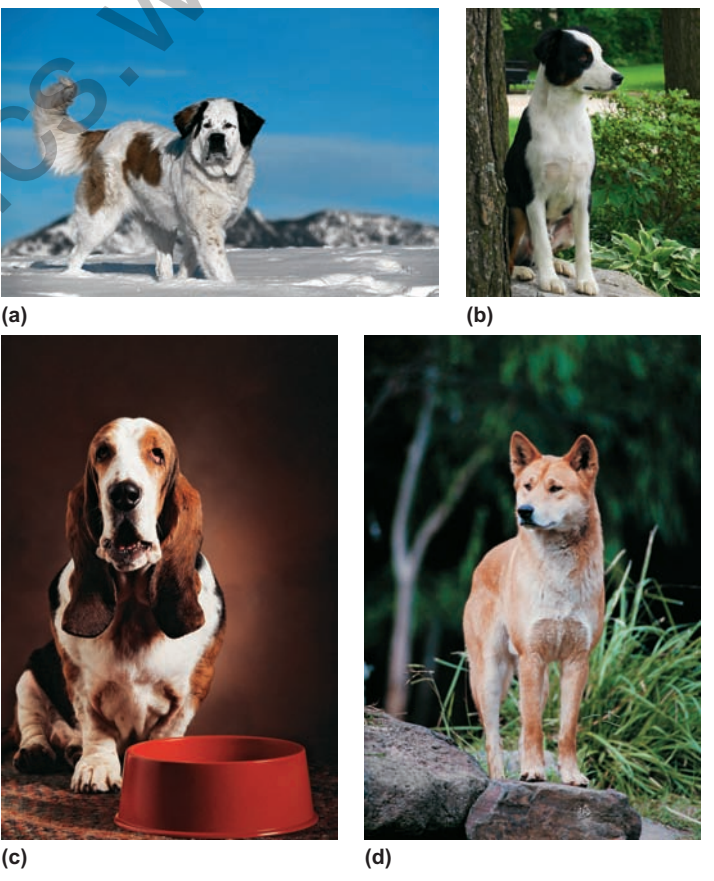
Scientists must rely on a variety of ways to identify species, because they cannot test every individual by breeding it with another to see if those individuals will have fertile offspring. Furthermore, many kinds of organisms reproduce primarily by asexual means. Because organisms that reproduce exclusively by asexual methods do not exchange genes with any other individuals, they do not fit the *biological species* definition very well. The standards used to identify various species include differences in morphology, behavior, metabolism, and genes.

*Morphological characteristics* are commonly used to differentiate species. Scientists compare the physical features of living and fossilized organisms when trying to identify a species. The idea that organisms can be classified as a species based on their structural characteristics is called the *morphological species concept*. The members of a species usually look alike, and these similarities are useful but not foolproof ways to distinguish among species. For example, the males and females of many birds look different from one another yet are the same species (figure 12.13).

Many plants have color variations or differences in leaf shape that cause them to look quite different, although they are members of the same species. Within the species, the eastern gray squirrel has black members that many people assume to be a different species because they are so different in color. A good example of the genetic diversity within a species is demonstrated by the various breeds of dogs. A Saint Bernard does not look very much like a Basset hound; however, they are members of the same species (figure 12.14).

It is often difficult to distinguish among species based on morphology. For example, to most people all mosquitoes look

**FIGURE 12.13 Sexual Differences Within a Species**
Male and female mallard ducks show strikingly different physical features, yet they are of the same species.



(a) (b) (c) (d)

**FIGURE 12.14 Genetic Diversity in Dogs**
Although these four breeds of dogs look quite different, they all have the same number of chromosomes and are capable of interbreeding. Therefore, they are considered to be members of the same species. *(a)* Saint Bernard, *(b)* Australian Shepherd, *(c)*Basset hound, *(d)* Australian dingo. Because the extremes of these breeds rarely interbreed naturally, the question is "How long will it be before they are no longer the same species?"

alike, but there are many species. And, although most people think all zebras are members of the same species, there are actually three species of zebras. In some cases, experts must use detailed morphology traits, such as the vein structure in the wings of insects, to identify species.

*Differences in behavior* are also useful in identifying species. Some species of birds are very similar structurally but can be easily identified by differences in the nature of their songs. Because it is often difficult to distinguish among bacteria, fungi, and other microorganisms on the basis of structure, *metabolic differences* that result in the presence or absence of specific chemicals within the organism are often used to help distinguish among species. The use of *genetic differences*—the analysis of DNA—is an even more precise way of distinguishing one species from another. Technology has allowed genes in many organisms to be sequenced and comparisons to be made. In some situations, this line of evidence has revealed remarkable differences between organisms once considered to be of the same species. In other cases, it has shown extremely similar sequences in what are considered to be very different species.

Because scientists use many standards—morphological, behavioral, metabolic, and genetic differences—to identify various species and none of these standards is perfect, situations frequently exist wherein individuals of two recognized species interbreed to a certain degree. For example, dogs, coyotes, and wolves have long been considered separate species. Differences in behavior and social systems tend to prevent mating among these three species. Wolves compete with coyotes and kill them when they are encountered. However, natural dog-coyote, wolf-coyote, and wolf-dog hybrids occur and the young are fertile (How Science Works 12.1). In fact, people have purposely encouraged mating between dogs and wolves for a variety of reasons. It has been demonstrated that dogs are descendants of wolves that were domesticated, so it should not be surprising that mating between wolves and dogs is easy to accomplish.

The species concept is an attempt to define groups of organisms that are *reproductively isolated* and, therefore, constitute a distinct unit of evolution. Some species are completely isolated from other, closely related species and do fit the definition well; some have occasional exchanges of genetic material between species and do not fit the definition as well; and some groups interbreed so much that they must be considered distinct populations of the same species. In this book, we will use the term *species* as a population of reproductively isolated individuals, complete with the flaws and shortcomings of this definition, because it is a useful way to identify groups of organisms that have great genetic similarity and maintain a certain degree of genetic separateness from all similar organisms.

There is one other thing you need to be careful about when using the word *species*. It is both a singular and plural word, so you can talk about a single species or several species. The only way to tell how the word is being used is by assessing the context of the sentence.

### 12.6 CONCEPT REVIEW

16. List four techniques used to distinguish one species from another.
17. Explain why the terms *reproductively isolated* and *species* are related.

## 12.7 Human Population Genetics

Recall from earlier in this chapter that the human gene pool consists of a number of subgroups. The particular characteristics that set one group apart from another originated many thousands of years ago, before travel was as common as it is today, and we still associate certain physical features with certain geographic areas. Although there is much more movement of people and a mixing of racial and ethnic types today, people still tend to have children with others who are of the same social, racial, and economic background and who live in the same locality.

This non-random mate selection can sometimes bring together two individuals who have alleles that are relatively rare. Information about allele frequencies within specific human subpopulations can be very important to people who wish to know the probability of having children with certain harmful genetic abnormalities. This is important if both individuals are descended from a common ancestral tribal, ethnic, or religious group. For example, Tay-Sachs disease causes degeneration of the nervous system and the early death of children. Because it is caused by a recessive allele, both parents must pass on the allele to their child in order for the child to have the disease. By knowing the frequency of the allele in the backgrounds of both parents, the probability of their having a child with this disease can be determined.

Ashkenazi Jews have a higher frequency of the recessive allele for Tay-Sachs disease than do people of any other group of racial or social origin, and the Jewish population of New York City has a slightly higher frequency of this allele than does the worldwide population of Ashkenazi Jews (figure 12.15). Therefore, people with this background should be aware of the probability that they will have children who will develop Tay-Sachs disease, even though the allele is moving to populations other than the Ashkenazi.

Likewise, sickle-cell anemia is more common in people of specific African ancestry than in any other human subgroup. Because many black slaves were brought to this country from regions where sickle-cell anemia is common, African Americans should be aware that they might carry the allele for this type of defective hemoglobin. If they carry the allele, they should consider their chances of having children with this disease. These and other cases make it very important that trained *genetic counselors* have information about allele frequencies in specific human ethnic groups, so that they can help couples with genetics questions.

## HOW SCIENCE WORKS 12.1

# The Legal Implications of Defining a Species

The red wolf (*Canis rufus*) is an endangered species, so the U.S. Fish and Wildlife Service has instituted a captive breeding program to preserve the animal and reintroduce it to a suitable habitat in the southeastern United States, where it was common into the 1800s. Biologists have long known that red wolves hybridize with both the coyote (*Canis latrans*) and the gray wolf (*Canis lupus*), and many suspect that the red wolf is actually a hybrid between the gray wolf and coyote. Gray wolf-coyote hybrids are common in nature where one of the species is rare. Some have argued that the red wolf does not meet the definition of a species and should not be protected under the Endangered Species Act.

Museums have helped shed light on this situation by providing skulls of all three kinds of animals preserved in the early 1900s. It is known that, during the early 1900s, as the number of red wolves in the southeastern United States declined, they readily interbred with coyotes, which were very common (the gray wolf had been exterminated by the early 1900s). Some scientists believe that the skulls of the few remaining "red wolves" might not represent the true red wolf but a "red wolf" with many coyote characteristics. Studies of the structure of the skulls of red wolves, coyotes, and gray wolves show that the red wolves were recognizably different and intermediate in structure between coyotes and gray wolves. This supports the hypothesis that the red wolf is a distinct species.

DNA studies were performed using material from preserved red wolf pelts. The red wolf DNA was compared with coyote and gray wolf DNA. These studies showed that red wolves contain DNA sequences of both gray wolves and coyotes but do not appear to have distinct base sequences found only in the red wolf. This evidence supports the opinion that the red wolf is not a species but may be a population that resulted from hybridization between gray wolves and coyotes.

There is still no consensus on the status of the red wolf. Independent researchers disagree with one another and with Fish and Wildlife Service scientists, who have been responsible for developing and administering a captive breeding program and planning the reintroductions of the red wolf.

**(a) Gray wolf, *Canis lupus***

**(b) Red wolf, *Canis rufus***

**(c) Coyote, *Canis latrans***

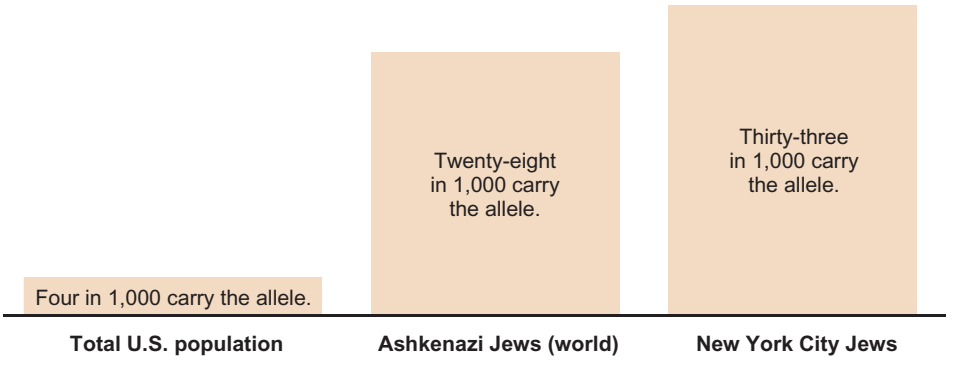



**FIGURE 12.15 The Frequency of the Tay-Sachs Allele**
The frequency of an allele can vary from one population to another. Genetic counselors use this information to advise people of their chances of having specific alleles and of passing them on to their children.

## 12.7 CONCEPT REVIEW

18. Give an example of a human population with a high frequency of a deleterious allele.
19. What is the role of a genetic counselor?

## 12.8 Ethics and Human Population Genetics

Misunderstanding the principles of heredity and population genetics has resulted in bad public policy. Often, when there is misunderstanding there is mistrust. Even today, many prejudices against certain genetic conditions persist.

Modern genetics had its start in 1900, with the rediscovery of the fundamental laws of inheritance proposed by Mendel. For the next 40 or 50 years, this rather simple understanding of genetics resulted in unreasonable expectations on the part of both scientists and laypeople. People generally assumed that much of what a person was in terms of structure, intelligence, and behavior was inherited. This led to the passage of *eugenics laws,* whose basic purpose was to eliminate "bad genes" from the human gene pool and encourage "good genes." These laws often prevented the marriage or permitted the sterilization of people who were "known" to have "bad genes" (figure 12.16). Often, these laws were promoted as a way to save money, because sterilization would prevent the birth of future "defectives" and, therefore, would reduce the need for expensive mental institutions and prisons. Some people used these laws to legitimize racism and promote prejudice.

The writers of eugenics laws (How Science Works 12.2) overestimated the importance of genetics and underestimated the significance of environmental factors such as disease and poor nutrition. They also overlooked the fact that many genetic abnormalities are caused by recessive alleles. In most cases, the negative effects of recessive alleles can be recognized only in homozygous individuals. Removing only the homozygous individuals from the gene pool would have little influence on the frequency of the "bad genes" in the population. Many recessive alleles would be masked by dominant alleles in heterozygous individuals and would continue to show up in future generations. In addition, we now know that most characteristics are not inherited in a simple dominant/recessive fashion; and that often many alleles at different loci cooperate in the production of a phenotypic

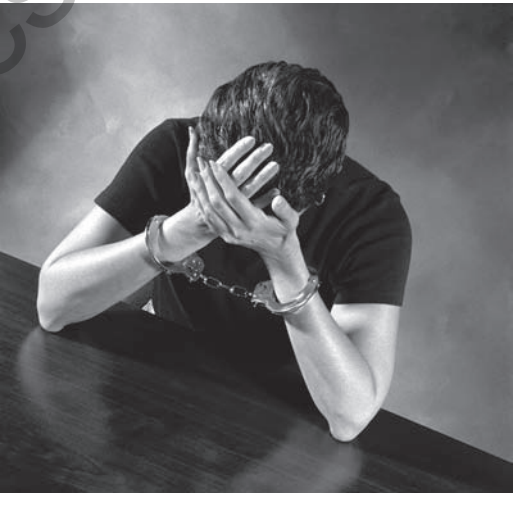


**FIGURE 12.16 A Eugenics Law**
**720.301 Sterilization of mental defectives; statement of policy**

Sec. 1. It is hereby declared to be the policy of the state to prevent the procreation and increase in number of feebleminded and insane persons, idiots, imbeciles, moral degenerates and sexual perverts, likely to become a menace to society or wards of the state. The provisions of this act are to be liberally construed to accomplish this purpose. As amended 1962, No. 160, § 1, Eff. March 28, 1963.

This state law was enacted in 1929 and is typical of many such laws passed during the 1920s and 1930s. A basic assumption of this law is that the conditions listed are inheritable; therefore, the sterilization of affected persons would decrease the frequency of these conditions. Prior to 1962, the law also included epileptics. It was repealed in 1974.

## HOW SCIENCE WORKS 12.2

# Bad Science: A Brief History of the Eugenics Movement

- **1885:** Francis Galton, cousin to Charles Darwin, proposes that human society could be improved "through better breeding." The term *eugenics* is coined; it is "the systematic elimination of undesirables to improve humanity." This would be accomplished by breeding those with "desirable" traits and preventing reproduction of those with "undesirable" traits. John Humphrey Noyes, an American sexual libertarian, molds the eugenics concept to justify polygamy: "While the good man will be limited by his conscience to what the law allows, the bad man, free from moral check, will distribute his seed beyond the legal limit."
- **1907:** Indiana is the first state to pass an involuntary sterilization law.
- **1919:** Charles B. Davenport, founder of Cold Springs Harbor Laboratory and of the Eugenics Record Office, "proves" that "pauperism" is inherited and "that being a naval officer is an inherited trait." He notes that the lack of women in the navy also "proves" that the gene is unique to males.
- **1920:** Davenport founds the American Eugenics Society. He sponsors "Fitter Families Contests," held at many state fairs around the country. The society persuades 20 state governments to authorize the sterilization of men and women in prisons and mental hospitals. The society also puts pressure on the federal government to restrict the immigration of "undesirable" races into the United States.
- **1927:** Oliver Wendel Holmes argues for the involuntary sterilization of Carrie S. Buck, an 18-year-old resident of the Virginia State Colony for Epileptics and Feeble-Minded. Buck is the first person to be selected for sterilization under the law. Buck is sterilized, even though it is later revealed that neither she nor her illegitimate daughter, Vivian, is feebleminded.
- **1931:** Involuntary sterilization measures are passed by 30 states.
- **1933–1941:** Nazi death camps, with the mass murder of Jews, Gypsies, Poles, and Russians, are established and run, resulting in the extermination of millions of people. According to the *New York Times* (August 29, 1935), "Adolf Hitler, . . . guided by the nation's anthropologists, eugenists and social philosophers, has been able to construct a comprehensive racial policy of population development and improvement. . . . It sets a pattern. . . . These ideas have met stout opposition in the Rousseauian social philosophy, . . . which bases . . . its whole social and political theory upon the patent fallacy of human equality. . . . Racial consanguinity occurs only through endogamous mating or interbreeding within racial stock . . . conditions under which racial groups of distinctly superior hereditary qualities . . . have emerged."
- **1972–1973:** Up to 4,000 sterilizations are still performed in Virginia alone, and the federal government estimates that 25,000 adults are sterilized nationwide.

1919 Charles B. Davenport, founder of Cold Springs Harbor Laboratory and of the Eugenics Record Office, "proved" that "*pauperism*" was inherited. Also "proved that being a naval officer is an inherited trait." He noted that the lack of women in the navy also "proved" that the gene was unique to males.

- **1973:** Since March 1973, the American Eugenics Society has called itself The Society for the Study of Social Biology.
- **1987:** Eugenic sterilization of institutionalized retarded persons is still permissible in 19 states, but the laws are rarely carried out. Some states enact laws that forbid the sterilization of people in state institutions.
- **Present:** Some groups and individuals still hold to the concepts of eugenics, claiming that recent evidence "proves" that traits such as alcoholism, homosexuality, and schizophrenia are genetic and, therefore, should be eliminated from the population to "improve humanity." However, the movement lacks the organization and legal basis it held in the past. Modern genetic advances, such as genetic engineering techniques and the mapping of the human genome, allow the identification of individuals with specific genetic defects. Questions about who should have access to such information and how it may be used causes renewed interest in the eugenics debate.

characteristic. Furthermore, mutations occur constantly, adding new alleles to the mix. Usually, these new alleles are recessive and deleterious. Thus, essentially all individuals are carrying "bad genes."

Today, genetic diseases and the degree to which behavioral characteristics and intelligence are inherited are still important social and political issues. The emphasis, however, is on determining the specific method of inheritance and the specific biochemical pathways that result in what is currently labeled as insanity, lack of intelligence, or antisocial behavior. Although progress is slow, several genetic abnormalities have been "cured," or at least made tolerable, by medicines and control of the diet. For example, phenylketonuria (PKU) is a genetic disease caused by an abnormal biochemical pathway. If children with this condition are allowed to eat foods containing the amino acid phenylalanine, they will become mentally retarded. However, if phenylalanine is excluded from the diet, and certain other dietary adjustments are made, the children will develop normally. NutraSweet is a phenylalanine-based sweetener, so people with PKU must use caution when buying products that contain it. This abnormality can be diagnosed very easily by testing the urine of newborn infants.

Effective genetic counseling is the preferred method of dealing with genetic abnormalities. A person known to be a carrier of a "bad gene" can be told the likelihood of passing on that characteristic to the next generation before deciding whether or not to have children. In addition, *amniocentesis* (a medical procedure that samples amniotic fluid) and other tests make it possible to diagnose some genetic abnormalities early in pregnancy. If an abnormality is diagnosed, an abortion can be performed. Because abortion is unacceptable to some people, the counseling process must include a discussion of the facts about an abortion and the alternatives. Although at one time counselors often pushed people toward specific decisions, today it is considered inappropriate for counselors to be advocates; their role is to provide information that allows individuals to make the best decisions possible for them.

**Genetic counseling**

## 12.8 CONCEPT REVIEW

20. What is amniocentesis?
21. What were eugenics laws? List two facts about human genetics that the advocates of eugenics failed to consider.

## Summary

All organisms with similar genetic information and the potential to reproduce are members of the same species. A species usually consists of several local groups of individuals, known as populations. Groups of interbreeding organisms are members of a gene pool. Although individuals are limited in the number of alleles they can contain, within the population there may be many different kinds of alleles for a trait. Subpopulations may have different allele frequencies from one another.

Genetically distinct populations exist because local conditions may demand certain characteristics, founding populations may have had unrepresentative allele frequencies, and barriers may prevent the free flow of genetic information from one locality to another. Distinguishable subpopulations are known as subspecies, varieties, strains, breeds, or races.

Genetic diversity is generated by mutations, which can introduce new alleles; sexual reproduction, which can generate new genetic combinations; and migration, which can subtract genetic information from, or add genetic information to, a local population. The size of the population is also important, because small populations have reduced genetic diversity.

A knowledge of population genetics is useful for plant and animal breeders and for people who specialize in genetic counseling. The genetic diversity of domesticated plants and animals has been reduced as a result of striving to produce high frequencies of valuable alleles. Clones and intraspecific hybrids are examples. Understanding allele frequencies and how they differ in various populations sheds light on why certain alleles are common in some human populations. Such understanding is also valuable in counseling members of populations with high frequencies of alleles that are relatively rare in the general population.

## Key Terms

*Use the interactive flash cards on the* ***Concepts in Biology****, 14/e website to help you learn the meaning of these terms.*

allele frequency 250
biological species concept 249
founder effect 254
gene frequency 250
gene pool 248

genetic bottleneck 255
genetic diversity 252
genetic drift 254
interspecific hybrids 257
intraspecific hybrids 257
monoculture 258
population 248
population genetics 248
species 248
subspecies (breeds, varieties, strains, races) 251

## Basic Review

1. A(n) _____ is all the individuals of the same kind of organism found within a specified geographic region and time.
2. A(n) _____ is all the alleles of all the individuals in a population.
   a. gene pool
   b. population
   c. allele pool
   d. clone
3. Which of the following does not belong?
   a. subspecies
   b. breed
   c. variety
   d. culture
4. Which of the following is a reason that genetically distinct populations exits?
   a. adaptation
   b. the founder effect
   c. cloning
   d. All of the above are correct.
5. Genetic diversity in domesticated plants and animals is affected by
   a. selective breeding.
   b. genetic engineering.
   c. cloning.
   d. All of the above are correct.
6. Morphological, behavioral, metabolic, and genetic differences are all important
   a. standards used to identify species.
   b. ways of generating genetic diversity in a population.
   c. reasons for the existence of gene pools.
   d. sources of mutation.
7. A(n) _____ is a form of genetic drift in which there is a sharp reduction in population size due to a chance event that results in a reduction in genetic diversity in subsequent generations.
8. The organisms that are produced by the controlled breeding of separate varieties of the same species are often referred to as
   a. intraspecific hybrids.
   b. interspecific hybrids.
   c. mutants.
   d. clones.
9. Which of the following causes degeneration of the nervous system and the early death of children?
   a. sickle-cell anemia
   b. Tay-Sachs disease
   c. PKU
   d. eugenics
10. _____ is the term used to describe genetic differences among members of a population.
11. The basic purpose of _____ _____ is to eliminate "bad genes" from the human gene pool and encourage "good genes."
12. If two organisms look different, it means that they are members of different species. (T/F)
13. A "zorse" is the result of breeding between a zebra and a horse and
   a. is an example of an interspecies hybrid.
   b. will no doubt be the beginning of a whole new species.
   c. only happens in zoos.
   d. is an example of intraspecies breeding.
14. Grape plants are grafted onto the stems of already-existing plants to generate more grape plants. This is also known as _____.
15. As a population decreases in size, it is most likely that
   a. genetic diversity will decrease.
   b. extinction is more likely.
   c. the gene pool also decreases in size.
   d. All of the above are true.

**Answers**
1. population 2. a 3. d 4. d 5. d 6. a 7. genetic bottleneck 8. a 9. b 10. genetic diversity 11. eugenics laws 12. F 13. a 14. cloning 15. d

## Thinking Critically

**Is GINA on Your Side?**

"The Genetic Information Nondiscrimination Act of 2008 (Pub.L. 110–233, 122 Stat. 881, enacted May 21, 2008, GINA), is an Act of Congress in the United States designed to prohibit the improper use of genetic information in health insurance and employment. The Act prohibits group health plans and health insurers from denying coverage to a healthy individual or charging that person higher premiums based solely on a genetic predisposition to developing a disease in the future. The legislation also bars employers from using individuals' genetic information when making hiring, firing, job placement, or promotion decisions."

Did you know about his law? What biological information led to the introduction of this bill? On what basis might a person have voted no on this bill? From your perspective, under what kinds of circumstances might a person file suit under this law?

CHAPTER 13

# Evolution and Natural Selection

## CHAPTER OUTLINE



## Study Reveals Information on Human Diversity and Evolution

*To understand the population genetics of any human population, it is necessary to understand Africa first.*

Africa is the birthplace of modern humans and, as a result, Africans have had more time to accumulate changes in their DNA. According to one researcher, ". . . genetically, Africans have been the most neglected and under-represented of any continental group, because the most diverse groups are often remote . . . and don't usually get to clinics."

That is until recently. An international team has analyzed nuclear DNA collected over a decade from 113 populations of Africans from across the continent. The team has found that Africans are descended from 14 ancestral populations, which often correlate with language and cultural groups. They found that all hunter-gatherers and pygmies in Africa today shared ancestors 35,000 years ago and that East Africa was the source of the great migration that populated the rest of the world. They also learned that African-American individuals, on average, have mixed ancestry from all over western Africa. This makes it difficult for African Americans to trace their roots to specific ethnic groups in Africa. The data also support research indicating that the source population for the out-of-Africa migration of modern humans came from east Africa near the Red Sea. These data give us raw material for understanding human evolution that has not been available until now.

- **How does separating groups of the same species into different geographic areas affect their diversity?**
- **What is the ultimate source of genetic variation among different populations?**
- **What environmental factors might have played roles in generating the genetic differences identified by this research?**

## Background Check

**Concepts you should already know to get the most out of this chapter:**

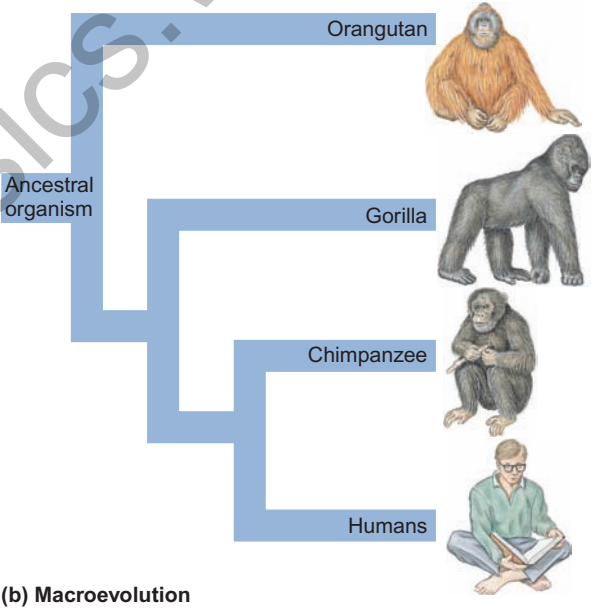
- Traits that make something alive (chapter 1)
- How an allele is involved in protein synthesis (chapter 8)
- The reasons why genetically different populations exist (chapter 12)
- How genetic diversity comes about (chapter 12)

## 13.1 The Scientific Concept of Evolution

People use the term *evolution* in many ways. We talk about the evolution of economies, fashion, and musical tastes. From a biological perspective, the word has a more specific meaning. **Evolution** is a change in the frequency of genetically determined characteristics within a population over time. Evolution can be looked at from two points of view. *Microevolution* occurs when there are minor differences in *allele frequency* between populations of the same species, as when scientists examine genetic differences between subspecies. *Macroevolution* occurs when there are major differences that have occurred over long periods that have resulted in so much genetic change that new *kinds of species* are produced (figure 13.1).

Regardless of the perspective, the ways these differences are brought about are basically the same. The focus of this chapter is on the processes that result in microevolutionary change. Chapter 14 focuses on processes that lead to macroevolutionary change—that is, the development of new species. Evolution, from both perspectives, involves changes in characteristics and the genetic information that produces these characteristics over many generations (Outlooks 13.1).

(a) Microevolution




(b) Macroevolution

**FIGURE 13.1 Microevolution and Macroevolution**

*(a)* Microevolution occurs when gene frequencies change within the gene pool of a species. Different populations of peppered moths show different gene frequencies based on the color of the bark of the trees they rest on. Black moths are conspicuous on light-colored trunks and light moths are conspicuous on dark colored trunks. Predation by birds removes more of the conspicuous moths and leads to the different gene frequencies in the two populations. These are relatively minor changes, compared with macroevolution changes, which result in new species from common ancestors. *(b)* The macroevolutionary pattern shown here may require tens of millions of years to occur and results in the formation of organisms that are so different that they are unable to interbreed.

## OUTLOOKS 13.1

### Common Misconceptions About the Theory of Evolution

1. **Evolution happened only in the past and is not occurring today.** In fact, there is much evidence that changes in the frequency of alleles are occurring in the populations of current species (e.g., antibiotic resistance, pesticide resistance).
2. **Evolution has a predetermined goal, or "it was meant to be."** Natural selection selects the organisms that best fit the current environment. As the environment changes, so do the characteristics that have value. Random events, such as changes in sea level, major changes in climate, volcanic eruptions, earthquakes, and collisions with asteroids, have had major influences on the subsequent natural selection and evolution.
3. **Changes in the environment cause the mutations that are needed to survive under the new environmental conditions.** Mutations are random events and are not necessarily adaptive. However, mutations that were originally detrimental or neutral may have greater value after the environment changes. The genetic information does not change, but the environmental conditions do. In some cases, the mutation rate may increase, or there may be more frequent genetic exchanges between individuals when the environment changes, but the mutations are still random. They are not directed toward a particular goal.
4. **Individual organisms evolve.** Individuals are stuck with the genes they have inherited from their parents. Although individuals may adapt by changing their behavior or physiology, they cannot evolve; only populations can change gene frequencies.

5. **Many of the current species can be shown to be derived from other present-day species (e.g., apes gave rise to humans).** There are few examples in which it can be demonstrated that one current species gave rise to another. Apes did not become humans, but apes and humans had a common ancestor several million years ago.
6. **Alleles that are valuable to an organism's survival become dominant.** An allele that is valuable may be either dominant or recessive. However, if it has a high value for survival, it will become *common* (more frequent). Commonness has nothing to do with dominance and recessiveness.

### 13.1 CONCEPT REVIEW

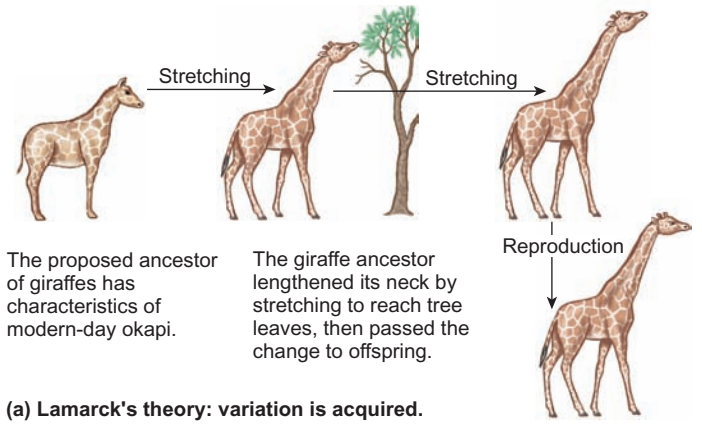
1. Describe the biological meaning of the word *evolution*.

## 13.2 The Development of Evolutionary Thought

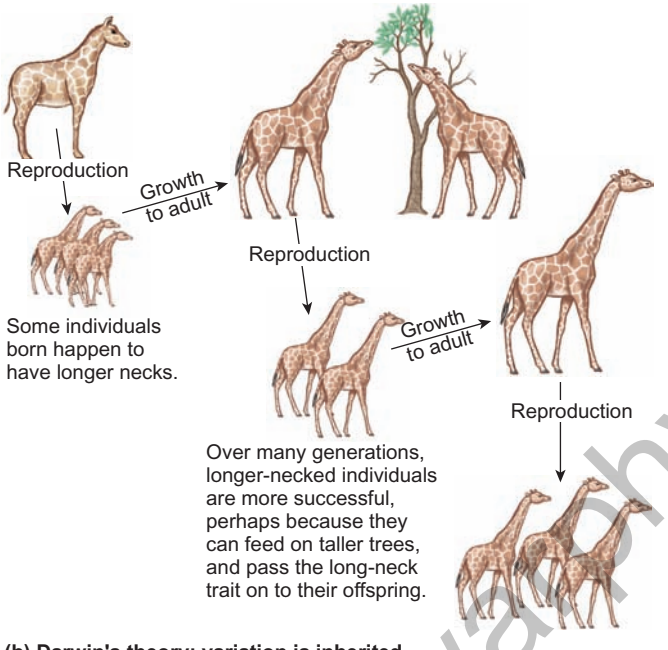
For centuries, people believed that the various species of plants and animals were unchanged from the time of their creation. Although today we know this is not true, we can understand why people may have thought it was true. Because they knew nothing about DNA, meiosis, genetics, or population genetics, they did not have the tools to examine the genetic nature of species. Furthermore, the process of evolution is so slow that the results of evolution are usually not recognized during a human lifetime. It is even difficult for modern scientists to recognize this slow change in many kinds of organisms.

### Early Thinking About Evolution

In the mid-1700s, Georges-Louis Buffon, a French naturalist, wondered if animals underwent change (evolved) over time. After all, if animals didn't change, they would stay the same, and it was becoming clear from the study of fossils that changes had occurred. However, Buffon didn't come up with any suggestions on how such changes might come about. In 1809, Jean-Baptiste de Lamarck, a student of Buffon's, suggested a process by which evolution might occur. He proposed that *acquired characteristics* were transmitted to offspring. **Acquired characteristics** are traits gained during an organism's life and not determined genetically. For example, he proposed that giraffes originally had short necks but because they constantly stretched their necks to get food, their necks got slightly longer (figure 13.2). When these giraffes reproduced, their offspring acquired their parents' longer necks. Because the offspring also stretched to eat, the third generation ended up with even longer necks. And so Lamarck's story was thought to explain why the giraffes we see today have long necks. Although we




**(a) Lamarck's theory: variation is acquired.**




**(b) Darwin's theory: variation is inherited.**

**FIGURE 13.2 The Contrasting Ideas of Lamarck and the Darwin-Wallace Theory**
*(a)* Lamarck thought that acquired characteristics could be passed on to the next generation. Therefore, he postulated that, as giraffes stretched their necks to get food, their necks got slightly longer. This characteristic was passed on to the next generation, which would have longer necks. *(b)* The Darwin-Wallace theory states that there is variation within the population and that those with longer necks are more likely to survive and reproduce, passing on their genes for long necks to the next generation.

now know Lamarck's theory was wrong (because acquired characteristics are not inherited), it stimulated further thought as to how evolution might occur. From the mid-1700s to the mid-1800s, lively arguments continued about the possibility of evolutionary change. Some, like Lamarck and others, thought that change did take place; many others said that it was not even possible. It was the thinking of two English scientists that finally provided a mechanism for explaining how evolution occurs.

## The Theory of Natural Selection

In 1858, Charles Darwin and Alfred Wallace suggested the theory of *natural selection* as a mechanism for evolution. The **theory of natural selection** is the idea that some individuals whose genetic combinations favor life in their surroundings are more likely to survive, reproduce, and pass on their genes to the next generation than are individuals who have unfavorable genetic combinations. This theory was clearly set forth in 1859 by Darwin in his book *On the Origin of Species by Means of Natural Selection, or the Preservation of Favored Races in the Struggle for Life* (How Science Works 13.1).

**Charles Darwin circa 1830**

**Alfred Wallace circa 1900**

The theory of natural selection is based on the following assumptions about the nature of living things:

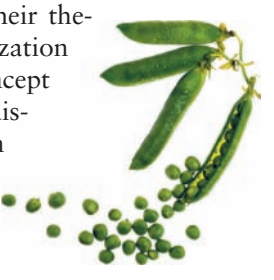
1. All organisms produce more offspring than can survive.
2. No two organisms are exactly alike.
3. Among organisms, there is a constant struggle for survival.
4. Individuals that possess favorable characteristics for their environment have a higher rate of survival and produce more offspring.
5. Favorable characteristics become more common in the species, and unfavorable characteristics are lost.

Using these assumptions, the Darwin-Wallace theory of evolution by natural selection offers a different explanation for the development of long necks in giraffes (figure 13.2b):

1. In each generation, more giraffes would be born than the food supply could support.
2. In each generation, some giraffes would inherit longer necks, and some would inherit shorter necks.
3. All giraffes would compete for the same food sources.
4. Giraffes with longer necks would obtain more food, have a higher survival rate, and produce more offspring.
5. As a result, succeeding generations would show an increase in the number of individuals with longer necks.

## Modern Interpretations of Natural Selection

The logic of the Darwin-Wallace theory of evolution by natural selection seems simple and obvious today, but at the time Darwin and Wallace proposed their theory, the processes of meiosis and fertilization were poorly understood, and the concept of the gene was only beginning to be discussed. Nearly 50 years after Darwin and Wallace suggested their theory, the rediscovery of the work a monk, Gregor Mendel (see chapter 10) provided an explanation for how characteristics could be transmitted from



**Garden Peas**

## HOW SCIENCE WORKS 13.1

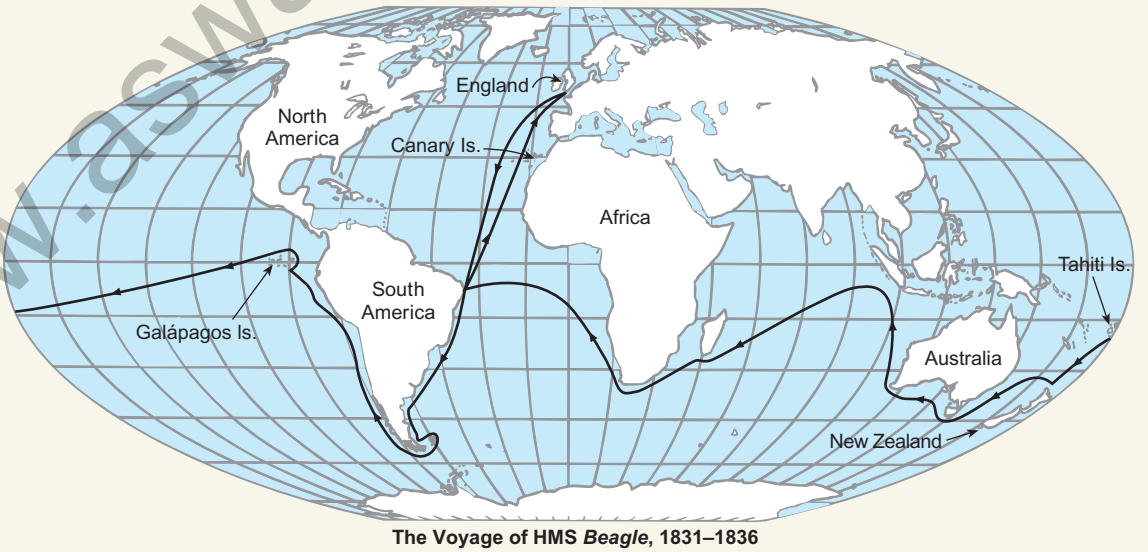
# The Voyage of HMS *Beagle*, 1831–1836

Probably the most significant event in Charles Darwin's life was his opportunity to sail on the British survey ship *Beagle*. Surveys were common at that time; they helped refine maps and chart hazards to shipping. Darwin was 22 years old and probably would not have had the opportunity, had his uncle not persuaded Darwin's father to allow him to take the voyage. Darwin was to be a gentleman naturalist and companion to the ship's captain, Robert Fitzroy. When the official naturalist left the ship and returned to England, Darwin replaced him and became the official naturalist for the voyage. The appointment was not a paid position.

The voyage of the *Beagle* lasted nearly 5 years. During the trip, the ship visited South America, the Galápagos Islands, Australia, and many Pacific Islands. The *Beagle*'s entire route is shown on the accompanying map. Darwin suffered greatly from seasickness and, perhaps because of it, made extensive journeys by mule and on foot some distance inland from wherever the *Beagle* happened to be at anchor. These inland trips gave Darwin the opportunity to make many of his observations. His experience was unique for a man so young and very difficult to duplicate because of the slow methods of travel used at that time.

Charles Darwin set forth on a sailing vessel similar to this, the HMS *Beagle*, in 1831 at the age of 22.

Although many people had seen the places that Darwin visited, never before had a student of nature collected volumes of information on them. Also, most other people who had visited these faraway places were military men or adventurers who did not recognize the significance of what they saw. Darwin's notebooks included information on plants, animals, rocks, geography, climate, and the native peoples he encountered. The natural history notes he took during the voyage served as a vast storehouse of information, which he used in his writings for the rest of his life. Because Darwin was wealthy, he did not need to work to earn a living and could devote a good deal of his time to the further study of natural history and the analysis of his notes. He was a semi-invalid during much of his later life. Many people think his ill health was caused by a tropical disease he contracted during the voyage of the *Beagle*. As a result of his experiences, he wrote several volumes detailing the events of the voyage, which were first published in 1839 in conjunction with other information related to the voyage. His volumes were revised several times and eventually were entitled *The Voyage of the Beagle*. He also wrote books on barnacles, the formation of coral reefs, how volcanoes might have been involved in reef formation, and finally *On the Origin of Species*. This last book, written 23 years after his return from the voyage, changed biological thinking for all time.




**The Voyage of HMS *Beagle*, 1831–1836**

0 1,000 2,000 3,000

Equatorial scale of miles

one generation to the next. Mendel's concepts of the gene explained how traits could be passed from one generation to the next. It also provided the first step in understanding mutations, gene flow, and the significance of reproductive isolation. All of these ideas are interwoven into the modern concept of evolution. If we update the five basic ideas from the thinking of Darwin and Wallace, they might look something like the following:

1. An organism's ability to overreproduce results in surplus organisms.
2. Because of mutation, new, genetically determined traits enter the gene pool. Because of sexual reproduction, involving meiosis and fertilization, new genetic combinations are present in every generation. These processes are so powerful that each individual in a sexually reproducing population is genetically unique. The genetic information present is expressed in the phenotype of the organism.
3. Resources, such as food, soil nutrients, water, mates, and nest materials, are in short supply, so some individuals do without. Other environmental factors, such as disease organisms, predators, and helpful partnerships with other species, also affect survival. All the specific environmental factors that affect survival by favoring certain characteristics are called **selecting agents**.
4. Selecting agents favor individuals with the best combination of alleles—that is, those individuals are more likely to survive and reproduce, passing on more of their genes to the next generation. An organism is selected against if it has fewer offspring than other individuals that have a more favorable combination of alleles. The organism does not need to die to be selected against.
5. Therefore, alleles or allele combinations that produce characteristics favorable to survival become more common in the population and, on the average, the members of the species will be better adapted to their environment.

Evolution results when there are changes in allele frequency in a population. Recall that individual organisms cannot evolve—only populations can. Although evolution is a population process, the mechanisms that bring it about operate at the level of the individual.

Recall that a theory is a well-established generalization supported by many kinds of evidence. The theory of natural selection was first proposed by Charles Darwin and Alfred Wallace. Since the time it was first proposed, the theory of natural selection has been subjected to countless tests yet remains the core concept for explaining how evolution occurs.

### 13.2 CONCEPT REVIEW

2. Why has Lamarck's theory been rejected?
3. List five assumptions about the nature of living things that support the concept of evolution by natural selection.

## 13.3 The Role of Natural Selection in Evolution

Natural selection is a primary process that brings about evolution by selecting which individuals will survive, reproduce, and pass on their genes to the next generation. These processes do not affect genes directly but do so indirectly by selecting individuals for success based on the characteristics an individual displays. Recall that the characteristics displayed by an organism (phenotype) are related to the genes possessed by the organism (genotype). By affecting the reproductive success of individuals, natural selection affects allele frequencies within the population. That change in allele frequency is evolution.

Road Kill Fox

Three factors work together to determine how a species changes over time: *environmental factors* that affect organisms, *sexual reproduction* among the individuals in the gene pool, and the amount of *genetic diversity* within the gene pool. In general, the reproductive success of any individual within a population is determined by how well an individual's characteristics match the demands of the environment in which it lives. **Fitness** is the success of an organism in passing on its genes to the next generation, compared with other members of its population. Just because an organism reproduces doesn't make it "fit." It can be fit only in comparison with others. Individuals whose characteristics enable them to survive and reproduce better than others in their environment have greater fitness (Outlooks 13.2).

Genetic diversity is important because a large gene pool with great genetic diversity is more likely to contain genetic combinations that allow some individuals to adapt to a changing environment. The characteristics of an organism are

## OUTLOOKS 13.2

### Genetic Diversity and Health Care

People turn to their healthcare providers when they experience a medical problem, whether it is the result of an accident, infection, or some abnormality. In many large cities, the emergency rooms (ERs) of large hospitals have become a substitute for a visit to a physician's office or a neighborhood clinic. Medical facilities are thought of as places where everyone always gets better and no one gets sick. However, 2 million people a year get bacterial infections while they are being treated in hospitals as patients. An estimated 90,000 people die from these infections each year.

Many people do not realize that the hospital is a place where patients who have not been able to have their infections resolved by home care bring all of those nasty microbes. Studies have shown that the farther away you are from the hospital, the less dangerous the microbes. What makes this situation worse is the fact that these bacteria are undergoing genetic changes and becoming more unbeatable. Populations of hospital microbes contain mutations that protect them from specific antibiotics—that is, they are antibiotic resistant. If resistant microbes are transmitted, the infected person will find the infection even harder to control. For example, methicillin-resistant *Staphylococcus aureus* (MRSA) was responsible for over 94,000 potentially fatal infections and nearly 19,000 deaths in the United States in 2005. Eighty-five percent of these deaths were associated with healthcare settings.


not just its structural traits. Behavioral, biochemical, and metabolic traits are also important. Scientists often use behavior, DNA differences, and other chemical differences to assess evolutionary relationships among existing organisms. However, when looking at extinct species, scientists are usually confined to using structural characteristics to guide their thinking.

### 13.3 CONCEPT REVIEW

4. Define *natural selection*.
5. What is fitness, and how is it related to reproduction?

## 13.4 Common Misunderstandings About Natural Selection

There are several common misinterpretations about the process of natural selection. The first involves the phrase "survival of the fittest." Individual survival is certainly important, because those that do not survive will not reproduce. However, the more important factor is the number of descendants an organism leaves. An organism that has survived for hundreds of years but has not reproduced has not contributed any of its genes to the next generation and, so, has been selected against. Therefore, the key to being the fittest is not survival alone but, rather, survival and reproduction of the more fit organisms.

Second, the phrase "struggle for life" in the title of Darwin's book does not necessarily refer to open conflict and fighting. It is usually much more subtle than that. When a resource, such as nesting material, water, sunlight, or food, is in short supply, some individuals survive and reproduce more effectively than others. For example, many kinds of birds require holes in trees as nesting places (figure 13.3). If these are in short supply, some birds are fortunate and find a top-quality nesting site, others occupy less suitable holes, and some do not find any. There may or may not be fighting for the possession of a site. If a site is already occupied, a bird may simply fly away and look for other suitable but less valuable sites. Those that successfully occupy good nesting sites will be much more successful in raising young than will those that must occupy poor sites or those that do not find any.

Similarly, on a forest floor where there is little sunlight, some small plants may grow fast and obtain light while shading out plants that grow more slowly. The struggle for life in this instance involves a subtle difference in the rate at which the plants grow. But the plants are, indeed, engaged in a struggle, and a superior growth rate is the weapon for survival.

**FIGURE 13.3 Tree Holes as Nesting Sites**
Many kinds of birds, such as this Gilded Flicker (*Colaptes chrysoides*), nest in holes in trees or other plants such as this Saguaro cactus. If such nesting sites are not available, they may not be able to breed. Many people build birdhouses that provide artificial tree holes to encourage birds to nest near their homes.

A third common misunderstanding involves the significance of phenotypic characteristics that are gained during the life of an organism but are not genetically determined. Although such acquired characteristics may be important to an individual's success, they are not genetically determined and cannot be passed on to future generations through sexual reproduction. Therefore, acquired characteristics are not important to the processes of natural selection. Consider an excellent golfer's skill. Although he or she may have inherited the physical characteristics of good eyesight, strength, and muscular coordination that are beneficial to a golfer, the ability to play a good round of golf is acquired through practice, not through genes. An excellent golfer's offspring will not automatically be excellent golfers. They might inherit some of the genetically determined physical characteristics necessary to become excellent golfers, however (figure 13.4).

Humans desire a specific set of characteristics in our domesticated animals. For example, the standard for the breed of dog known as boxers is for them to have short tails. However, the alleles for short tails are rare in this breed. Consequently, their tails are amputated—a procedure called docking. Similarly, most lambs' tails are amputated. These acquired characteristics are not passed on to the next generation. Removing the tails of these animals does not remove the genetic information for tail production from their genomes, and each generation of puppies and lambs is born with long tails.

**FIGURE 13.4 Acquired Characteristics**
The ability to play an outstanding game of golf is learned through long hours of practice. The golf skills acquired by practice cannot be passed on genetically to a person's offspring.

A fourth common misconception involves understanding the relationship between the mechanism of natural selection and the outcomes of the selection process. Although the effects of natural selection appear at the population level, the actual selecting events take place, one at a time, at the level of the individual organism.

### 13.4 CONCEPT REVIEW

6. Why are acquired characteristics of little interest to evolutionary biologists?
7. In what way are the phrases "survival of the fittest" and "struggle for existence" correct? In what ways are they misleading?

## 13.5 What Influences Natural Selection?

Now that you have a basic understanding of how natural selection works, we can look in more detail at the factors that influence it. *Genetic diversity* within a species, the degree of *genetic expression,* and the ability of most species to *reproduce excess offspring* all exert an influence on the process of natural selection.

### The Mechanisms That Affect Genetic Diversity

For natural selection to occur there must be genetic differences among the individuals of an interbreeding population of organisms. Consider what happens in a population of genetically identical organisms. In this case, it does not matter

which individuals reproduce, because the same genes will be passed on to the next generation and natural selection cannot occur. However, when genetic differences exist among individuals in a population and these differences affect fitness, natural selection can take place. Therefore, it is important to identify the processes that generate genetic diversity within a population. Genetic diversity within a population is generated by the *mutation* and *migration* of organisms and by *sexual reproduction* and *genetic recombination*.

## Mutation and Migration

**Spontaneous mutations** are changes in DNA that cannot be tied to a particular factor. Mutations may alter existing genes, resulting in the introduction of entirely new genetic information into a gene pool. It is suspected that cosmic radiation or naturally occurring mutagenic chemicals might be the cause of many of these mutations. Subjecting organisms to high levels of radiation or to certain chemicals increases the rate at which mutations occur. It is for this reason that people who are exposed to mutagenic chemicals or radiation take special safety precautions.

**Protective Lead Vest for Dental X-ray**

Naturally occurring mutation rates are low. The odds of a gene mutating are on the order of 1 in 100,000. Most of these mutations are harmful. Rarely does a mutation occur that is actually helpful. However, in populations of millions of individuals, each of whom has thousands of genes, over thousands of generations it is quite possible that a new, beneficial piece of genetic information will come about as a result of mutation. Remember that every allele originated as a modification of a previously existing piece of DNA. For example, the allele for blue eyes may be a mutated brown-eye allele, or blond hair may have originated as a mutated brown-hair allele. In a species such as corn (*Zea mays*), there are many different alleles for seed color. Each probably originated as a mutation. Thus, mutations have been very important in introducing genetic material into species over time.

For mutations to be important in the evolution of organisms, they must be in cells that give rise to gametes (eggs or sperm). Mutations in other cells, such as those in the skin or liver, will affect only those cells and will not be passed on to the next generation.

**Genetic Diversity in Corn**

Recall that migration is another way in which new genetic material can enter a population. When individuals migrate into a population from some other population, they may bring alleles that were rare or absent. Similarly, when individuals leave a population, they can remove certain alleles from the population.

## Sexual Reproduction and Genetic Recombination

Sexual reproduction is important in generating new genetic combinations in individuals. Although sexual reproduction does *not* generate new genetic information, it does allow for the mixing of genes into combinations that did not occur previously. Each individual entering a population by sexual reproduction carries a unique combination of genes—half donated by the mother and half donated by the father. During meiosis, unique combinations of alleles are generated in the gametes through crossing-over between homologous chromosomes and the independent assortment of nonhomologous chromosomes. This results in millions of possible genetic combinations in the gametes of any individual. When fertilization occurs, one of the millions of possible sperm unites with one of the millions of possible eggs, resulting in a genetically unique individual. This genetic mixing that occurs as a result of meiosis and fertilization is known as **genetic recombination**.

The new individual has a set of genes that is different from that of any other organism that ever existed. When genetic

recombination occurs, a new combination of alleles may give its bearer a selective advantage, leading to greater reproductive success.

Organisms that primarily use asexual reproduction do not benefit from genetic recombination. In most cases, however, when their life history is studied closely, it is apparent that they also can reproduce sexually at certain times. Organisms that reproduce exclusively by asexual methods are not able to generate new genetic combinations but still acquire new genetic information through mutation.

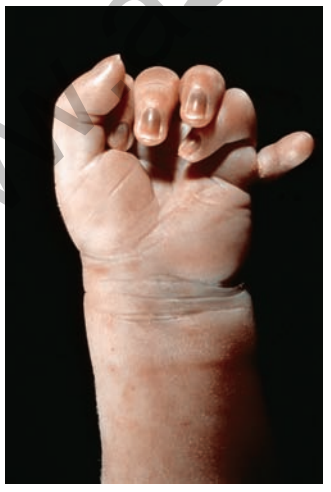
As scientists have learned more about the nature of species, it has become clear that genes can be moved from one organism to another that was considered to be a different species. In some cases, it appears that whole genomes can be added when the cells of two different species combine into one cell. This process of interspecific hybridization is another method by which a species can have new genes enter its population.

## The Role of Gene Expression

Even when genes are present, they do not always express themselves in the same way. *For genes to be selected for or against, they must be expressed in the phenotype of the individuals possessing them*. There are many cases of genetic characteristics being expressed to different degrees in different individuals. Often, the reason for this difference in expression is unknown.

### Degrees of Expression

**Penetrance** is a term used to describe how often an allele expresses itself. Some alleles have 100% penetrance; others express themselves only 80% of the time. For example, there is a dominant allele that causes people to have a stiff little finger. The expression of this trait results in the tendons being attached to the bones of the finger in such a way that the finger does not flex properly. This dominant allele does not express itself in every person who contains it; occasionally, parents who do not show the characteristic in their phenotype have children that show the characteristic. **Expressivity** is a term used to describe situations in which an allele is not expressed equally in all individuals who have it. An example of expressivity involves a dominant allele for six fingers or toes, a condition known as polydactyly. Some people with this allele have an extra finger on each hand; some have an extra finger on only one hand. Furthermore, some sixth fingers are well-formed with normal bones, whereas others are fleshy structures that lack bones.



**Polydactyly**

### Why Some Genes May Avoid Natural Selection

There are many reasons a specific allele may not feel the effects of natural selection. Some genetic characteristics can be expressed only during specific periods in the life of an organism. If an organism dies before the characteristic is expressed, it never has the opportunity to contribute to the overall fitness of the organism. Say, for example, a tree has genes for producing very attractive fruit. The attractive fruit is important because animals select the fruit for food and distribute the seeds as they travel. However, if the tree dies before it can reproduce, the characteristic may never be expressed. By contrast, genes such as those that contribute to heart disease or cancer usually have their effect late in a person's life. Because they were not expressed during the person's reproductive years, they were not selected against, because the person reproduced before the effects of the gene were apparent. Therefore, such genes are less likely to be selected against (eliminated from the population) than are those that express themselves early in life.

In addition, many genes require an environmental trigger to be expressed (i.e., they are epigenetic). If the trigger is not encountered, the gene never expresses itself. It is becoming clear that many kinds of human cancers are caused by the presence of genes that require an environmental trigger. Therefore, we try to identify triggers and prevent these negative genes from being turned on and causing disease.

When both dominant and recessive alleles are present for a characteristic, the recessive alleles must be present in a homozygous condition before they have an opportunity to express themselves. For example, the allele for albinism is recessive. There are people who carry this recessive allele but never express it, because it is masked by the dominant allele for normal pigmentation (figure 13.5).

Some genes have their expression hidden because the action of a completely unrelated gene is required before they can express themselves. The albino individual shown in figure 13.5 has alleles for dark skin and hair that will never have a chance to express themselves because of the presence of two alleles for albinism. The alleles for dark skin and hair can express themselves only if the person has the ability to produce pigment, and albinos lack that ability.

### Natural Selection Works on the Total Phenotype

Just because an organism has a "good" gene does not guarantee that it will be passed on. The organism may also have "bad" genes in combination with the good, and the "good" characteristics may be overshadowed by the "bad" characteristics. All individuals produced by sexual reproduction probably have

**FIGURE 13.5 Gene Expression**
Genes must be expressed to allow the environment to select for or against them. The recessive allele *c* for albinism shows itself only in individuals who are homozygous for the recessive characteristic. The man in this photo is an albino who has the genotype *cc*. The characteristic is absent in those who are homozygous dominant and is hidden in those who are heterozygous. The dark-skinned individuals could be either *Cc* or *CC*. However, because the albino individual cannot produce pigment, characteristics for dark skin and dark hair cannot be expressed.

**FIGURE 13.6 Reproductive Potential**
The ability of a population to reproduce greatly exceeds the number necessary to replace those who die. Here are some examples of the prodigious reproductive abilities of some species.

certain genetic characteristics that are extremely valuable for survival and others that are less valuable or harmful. However, natural selection operates on the total phenotype of the organism. Therefore, it is the combination of characteristics that is evaluated—not each characteristic individually. For example, fruit flies may show resistance to insecticides or lack of it, may have well-formed or shriveled wings, and may exhibit normal vision or blindness. An individual with insecticide resistance, shriveled wings, and normal vision has two good characteristics and one negative one, but it would not be as successful as an individual with insecticide resistance, normal wings, and normal vision.

## The Importance of Excess Reproduction

A successful organism reproduces at a rate in excess of that necessary to merely replace the parents when they die (figure 13.6). For example, geese have a life span of about 10 years; on average, a single pair can raise a brood of about eight young each year. If these two parent birds and all their offspring were to survive and reproduce at this rate for a 10-year period, there would be a total of 19,531,250 birds in the family.

However, the size of goose populations and most other populations remains relatively constant over time. Minor changes in number may occur but, if the environment remains constant, a population does not experience dramatic increases in size. A high death rate tends to offset the high reproductive rate and population size remains stable. But this is not a "static population." Although the total number of organisms in the species may remain constant, the individuals that make up the population change. It is this extravagant reproduction that provides the large surplus of genetically unique individuals that allows natural selection to take place.

If there are many genetically unique individuals within a population, it is highly probable that some individuals will survive to reproduce even if the environment changes somewhat, although the gene frequency of the population may be changed to some degree. For this to occur, members of the population must be eliminated in a non-random manner. Even if they are not eliminated, some may have greater reproductive success than others. The individuals with the greatest reproductive success will have more of their genetic information present in the next generation than will those that die or do not reproduce very successfully. Those that are the most successful at reproducing are those that are, for the most part, better suited to the environment.

### 13.5 CONCEPT REVIEW

8. What factors can contribute to diversity in the gene pool?
9. Why is over-reproduction necessary for evolution?
10. Why is sexual reproduction important to the process of natural selection?
11. How might a harmful allele remain in a gene pool for generations without being eliminated by natural selection?

## 13.6 The Processes That Drive Selection

Several mechanisms allow for the selection of certain individuals for successful reproduction. If predators must pursue swift prey organisms, the faster predators will be selected for, and the selecting agent is the swiftness of available prey. If predators must find prey that are slow but hard to see, the selecting agent is the camouflage coloration of the prey, and keen eyesight is selected for. If plants are eaten by insects, the production of toxic materials in the leaves is selected for. All selecting agents influence the likelihood that certain characteristics will be passed on to subsequent generations.

### Differential Survival

As stated previously, the phrase "survival of the fittest" is often associated with the theory of natural selection. Although this is recognized as an oversimplification of the concept, survival is an important factor in influencing the flow of genes to subsequent generations. If a population consists of a large number of genetically and phenotypically different individuals it is likely that some of them will possess characteristics that make their survival difficult. Therefore, they are likely to die early in life and not have an opportunity to pass on their genes to the next generation.

Charles Darwin described several species of ground finches on the Galápagos Islands (figure 13.7), and scientists have often used these birds in scientific studies of evolution. On one of the islands, scientists studied one of the species of seed-eating ground finches, *Geospiza fortis*. They measured the size of the animals and the size of their bills and related these characteristics to their survival. They found the following: During a drought, the birds ate the smaller, softer seeds more readily than the larger, harder seeds. As the birds consumed the more easily eaten seeds, only the larger, harder seeds remained. During the drought, finch mortality was extremely high. When scientists looked at ground finch mortality, they found that the larger birds with stronger, deeper bills survived better than the smaller birds with weaker, narrower bills. They also showed that the offspring of the survivors tended to show larger body and bill size as well. The lack of small, easily eaten seeds resulted in selection for larger birds with stronger bills, which could crack open larger, tougher seeds. Table 13.1 shows data on two of the parameters measured in this study.

As another example of how differential survival can lead to changed gene frequencies, consider what has happened to many insect populations as humans have subjected them to a variety of insecticides. Because there is genetic diversity within all species of insects, an insecticide that is used for the first time on a particular species kills all the exposed individuals that are genetically susceptible. However, individuals with slightly different genetic compositions and those not exposed may not be killed by the insecticide.

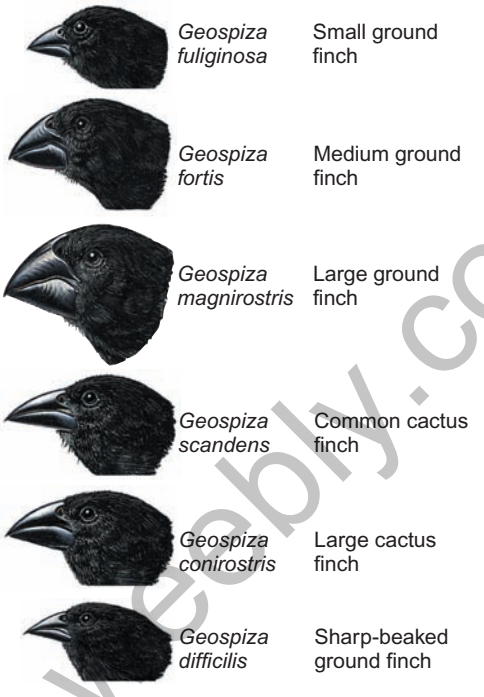



**FIGURE 13.7 A Sample of Darwin's Finches**
Ten species of ground and tree finches were described by Darwin. This figure shows six members of the genus *Geospiza*. The four species of ground finches are primarily seedeaters and use their bills to crush seeds. The two species of cactus finch primarily feed on the fruit and flowers of cactus plants.

**TABLE 13.1 Changes in Body Structure of *Geospiza fortis***

| | Before Drought | After Drought |
|---|---|---|
| *Average Body Weight* | 16.06 g | 17.13 g |
| *Average Bill Depth* | 9.21 mm | 9.70 mm |

Suppose that, in a population of a particular species of insect, 5% of the individuals have genes that make them resistant to a specific insecticide. The first application of the insecticide could, therefore, kill a majority of those exposed. However, tolerant individuals and those that escaped exposure would then constitute the remaining breeding population. This would mean that many more insects in the second generation would be tolerant. The second use of the insecticide on this population would not be as effective as the first. With continued use of the same insecticide, each generation would become more tolerant, because the individuals that were not tolerant were being eliminated and those that could tolerate the toxin passed on their genes for tolerance to their offspring.

Many species of insects produce a new generation each month. In organisms with a short generation time, 99% of the population could become resistant to an insecticide in just 5 years. As a result, the insecticide would no longer be

useful in controlling the species. As a new selecting agent (the insecticide) is introduced into the insect's environment, natural selection results in a change in the gene frequency of a population, so that most individuals are tolerant of the insecticide.

The same kind of selection process has occurred with herbicides. Within the past 50 years, many kinds of herbicides have been developed to control weeds in agricultural fields. After several years of use, a familiar pattern develops as more and more species of weeds show resistance to the herbicide. Figure 13.8 shows several kinds of herbicides and the number of weed species that have become resistant over time. In each weed species, there has been selection for the individuals that have the genetic information that allows them to tolerate the presence of the herbicide.

## Differential Reproductive Rates

Survival alone does not always ensure reproductive success. For a variety of reasons, some organisms are better able to use the available resources to produce offspring. If an individual leaves 100 offspring and another leaves only 2, the first organism has passed on more copies of its genetic information to the next generation than has the second. If we assume that all 102 offspring have similar survival rates, the first organism has been selected for, and its genes have become more common in the subsequent population.

Scientists have studied the gene frequencies for the height of clover plants. Two identical fields of clover were planted and cows were allowed to graze in one of them. The cows acted as a selecting agent by eating the taller plants first. These tall plants rarely got a chance to reproduce. Only the shorter plants flowered and produced seeds. After some time, seeds were collected from both the grazed and the ungrazed fields and grown in a greenhouse under identical conditions. The average height of the plants from the ungrazed field was compared with that of the plants from the grazed field. The seeds from the ungrazed field produced some tall, some short, but mostly medium-sized plants. However, the seeds from the grazed field produced many more short plants than medium or tall ones. The cows had selectively eaten the tall plants. Because the flowers are at the tip of the plant, the tall plants were less likely to successfully reproduce, even though they were able to survive grazing by cows.

## Differential Mate Choice—Sexual Selection

**Sexual selection** occurs within animal populations when some individuals are more likely to be chosen as mates than others. Obviously, those that are frequently chosen have more opportunities to pass on more copies of their genetic information than those that are rarely chosen. The characteristics of the more frequently chosen individuals may involve general characteristics, such as body size or aggressiveness, or specific, conspicuous characteristics attractive to the opposite sex.

For example, male red-winged blackbirds establish territories in cattail marshes, where females build their nests. A male will chase out all other males, but not females. Some blackbird territories are large and others are small; some males have none. Although it is possible for any male to mate, those that have no territory are least likely to mate. Those that defend large territories may have two or more females nesting in their territories and are very likely to mate with those females. It is unclear exactly why females choose one male's territory over another, but the fact is that some males are chosen as mates and others are not.

In other cases, it appears that females select males that display specific, conspicuous characteristics. Certain male birds, such as peacocks, have very conspicuous tail feathers

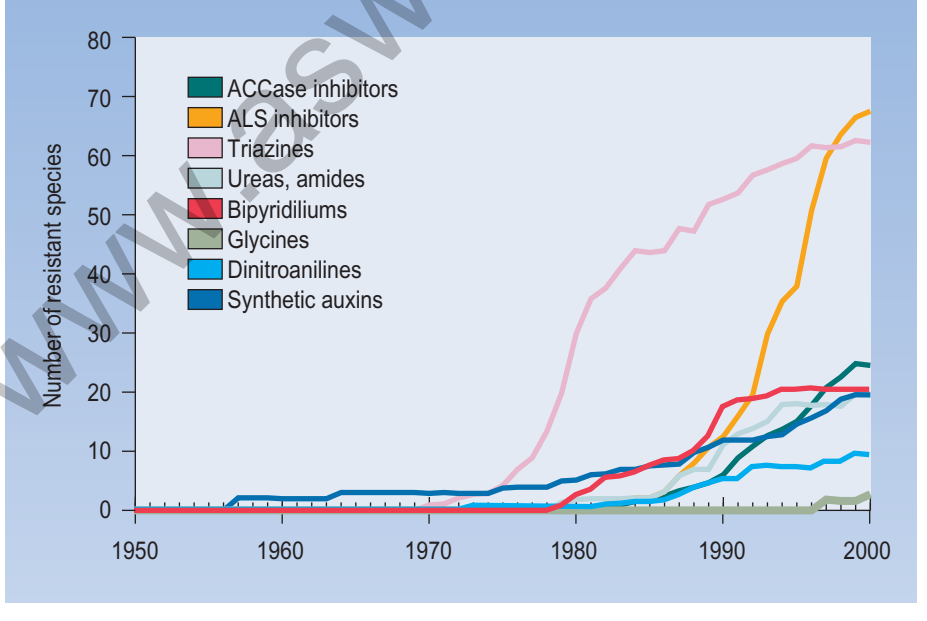
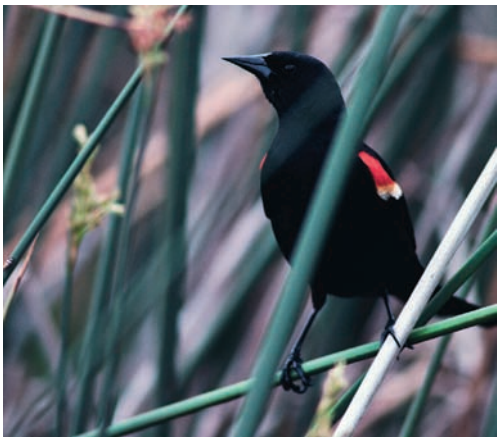



**FIGURE 13.8 Evolutionary Change**
Populations of weed plants that have been subjected repeatedly to herbicides often develop resistant populations. The individual weed plants that have been able to resist the effects of the herbicide have lived to reproduce and pass on their genes for resistance to their offspring; thus, resistant populations of weeds have developed.



Male Redwing Blackbird

(figure 13.9). Those with spectacular tails are more likely to mate and have offspring. Darwin was puzzled by such cases, because the large, conspicuous tail should have been a disadvantage to the bird. Long tails require energy to produce, make it more difficult to fly, and make it more likely that predators will capture the individual. The current theory that seeks to explain this paradox involves female choice. If the females have an innate (genetic) tendency to choose the most elaborately decorated males, genes that favor such plumage will be regularly passed on to the next generation.

### 13.6 CONCEPT REVIEW

12. List three factors that can lead to changed gene frequencies from one generation to the next.
13. Give two examples of selecting agents and explain how they operate.

## 13.7 Patterns of Selection

Understanding the nature of the environment is key to determining how natural selection will affect how a species will change. In general, three forms of selection have been identified: stabilizing selection, directional selection, and disruptive selection (figure 13.10).

### Stabilizing Selection

**Stabilizing selection** occurs when individuals at the extremes of the range of a characteristic are consistently selected against. This kind of selection is very common. If the environment is stable, most of the individuals show characteristics that are consistent with the demands of the environment. For example, for many kinds of animals, there is a range of color possibilities. Suppose a population of mice has mostly brown individuals and a few white or black ones. If the white or black individuals are more conspicuous and are consistently more likely to be discovered and killed by predators, the elimination of the extreme forms will result in a continued high frequency of the brown form. Many kinds of marine animals, such as horseshoe crabs and sharks, have remained unchanged for thousands of years. The marine environment is relatively constant and probably favors stabilizing selection.

### Directional Selection

**Directional selection** occurs when individuals at one extreme of the range of a characteristic are consistently selected for. This kind of selection often occurs when there is a consistent change in the environment in which the organism exists. For example, when a particular insecticide is introduced to control a certain species of pest insect, there is consistent selection for individuals that have alleles for resistance to the insecticide. Because of

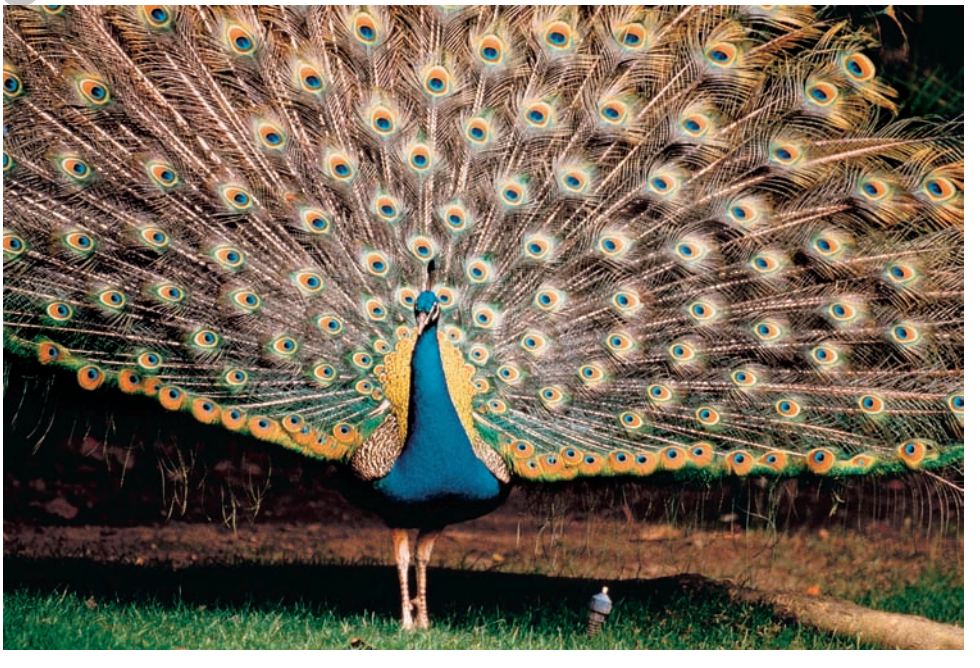


**FIGURE 13.9 Mate Selection**
In many animal species the males display very conspicuous characteristics that are attractive to females. Because the females choose the males they will mate with, those males with the most attractive characteristics will have more offspring and, in future generations, there will be a tendency to enhance the characteristic. With peacocks, those individuals with large colorful displays are more likely to mate.

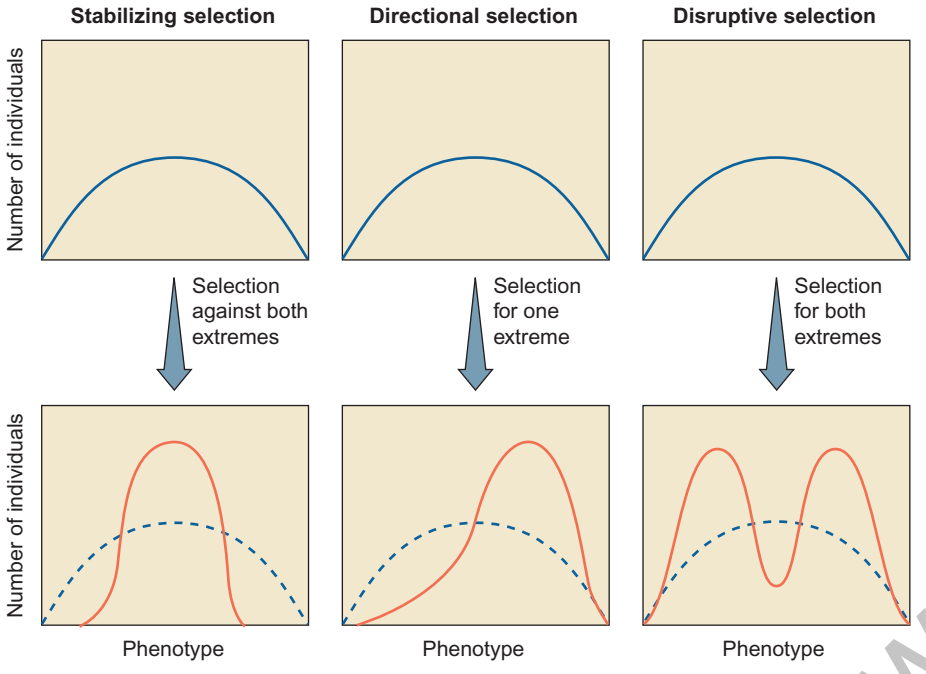



**FIGURE 13.10 Patterns of Selection**
In stabilizing selection, both extremes for a characteristic are selected against. Thus, gene frequencies do not change, and the original range of phenotypes is maintained. In directional selection, one extreme is selected for and the other extreme is selected against. This results in a shift in gene frequency and range of phenotype in a direction toward one extreme and away from the other. In disruptive selection, both extremes are selected for and the intermediate condition is selected against. This leads to the development of two distinct phenotypes with different gene frequencies.

this, there is a shift in the original allele frequency, from one in which the alleles for resistance to the insecticide were rare to one in which most of the population has the alleles for resistance. Similarly, changes in climate, such as long periods of drought, can consistently select for individuals that have characteristics that allow them to survive in the dryer environment, and a change in allele frequency can result. Recent evidence also shows changes resulting from periods of rapid climate warming. The Yukon red squirrel, pitcher-plant mosquito, and European blackcap warbler show genetically based shifts in the timing of their seasonal reproduction, dormancy, or migration.

## Disruptive Selection

**Disruptive selection** occurs when both extremes of a range for a characteristic are selected for and the intermediate condition is selected against. This kind of selection is likely to happen when there are sharp differences in the nature of the environment where the organisms live. For example, there are many kinds of insects that feed on the leaves of trees. Many of these insects have colors that match the leaves they feed on. Suppose the species of insect ranges in color from light green to dark green, and medium green is the most common. If a particular species of insect had some individuals that fed on plants with dark green leaves, whereas other individuals fed on plants with light green leaves, medium green insects could be selected against and the two extremes selected for, depending on the kind of plant they were feeding on.

### 13.7 CONCEPT REVIEW

14. Distinguish among stabilizing, directional, and disruptive selection.

## 13.8 Evolution Without Selection—Genetic Drift

Recall from chapter 12 that genetic drift is a significant change in the frequency of an allele that is not the result of natural selection. Gene-frequency differences that result from chance are more likely to occur in small populations than in large populations. Because they result from random events, such changes are not the result of natural selection or sexual selection. For example, a population of 10 organisms, of which 20% have curly hair and 80% have straight hair, is significantly changed by the death of 1 curly-haired individual. Often, the characteristics affected by genetic drift do not appear to have any adaptive value to the individuals in the population. However, in extremely small populations, vital genes may be lost.

Occasionally, a population has unusual colors, shapes, or behaviors, compared with other populations of the same species. Such unusual occurrences are associated with populations that started as a small founder population or those that have passed through a genetic bottleneck in the past. In large populations, any unusual shifts in gene frequency in one part of the population usually would be counteracted by reciprocal changes in other parts of the population. However, in small populations, the random distribution of genes to gametes may not reflect the percentages present in the population. For example, consider a situation in which there are 100 plants in a population and 10 have dominant alleles for patches of red color, whereas the others do not. If in those 10 plants the random formation of gametes resulted in no red alleles present in the gametes that were fertilized, the allele could be eliminated. Similarly, if all those plants with the red allele happened to be in a hollow that was subjected to low temperatures, they could be killed by a late frost and would not pass on their alleles to

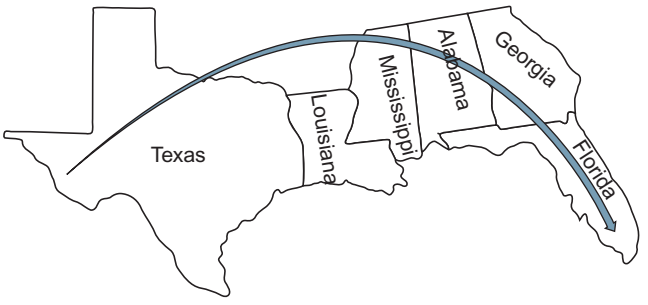



**FIGURE 13.11 The Florida Panther**
The Florida panther (also known as cougar, puma, mountain lion, or catamount) is confined to the Everglades at the southern tip of Florida. The population is small, is isolated from other populations of cougars, and prior to 1995 had lost about 50% of its genetic diversity. In 1995, 8 female cougars from Texas were introduced into the Everglades to increase the genetic diversity of the Florida panther population. The plan seems to be working. By 2002, the population had increased to about 80 adult cougars and the population had shown about 20% genes from the Texas subpopulation. Certain obvious characteristics, such as kinked tails and cowlicks, that were common in the pre-1995 population are now less common.

the next generation. Therefore, the allele would be lost, but the loss would not be the result of natural selection.

Consider the example of cougars in North America. Cougars require a wilderness setting for success. As Europeans settled the land over the past 200 years, the cougars were divided into small populations in the places where relatively undisturbed habitat still existed. The Florida panther is an isolated population of cougars found in the Everglades. The next nearest population of cougars is in Texas. Because the Florida panther is on the endangered species list, efforts have been made to ensure its continued existence in the Everglades. However, the population is small and studies show that it has little genetic diversity. A long period of isolation and a small population created conditions that led to this reduced genetic diversity. The accidental death of a few key individuals could have resulted in the loss of valuable genes from the population. The general health of the individuals in the population is poor and reproductive success is low. In 1995, wildlife biologists began a program of introducing individuals from the Texas population into the Florida population. The purpose of the program is to reintroduce the genetic diversity lost during the long period of isolation. The program appears to be working, because there has been an increase in genetic diversity within the population (figure 13.11).

### 13.8 CONCEPT REVIEW

15. Why is genetic drift more likely in small populations?
16. Give an example of genetic drift.

## 13.9 Gene-Frequency Studies and the Hardy-Weinberg Concept

In the early 1900s, an English mathematician, G. H. Hardy, and a German physician, Wilhelm Weinberg, recognized that it was possible to apply a simple mathematical relationship to the study of gene frequencies. Their basic idea was that, if certain conditions existed, gene frequencies would remain constant and the distribution of genotypes could be described by the relationship $p^2 + 2pq + q^2 = 1$, where $p^2$ represents the frequency of the homozygous dominant genotype, $2pq$ represents the frequency of the heterozygous genotype, and $q^2$ represents the frequency of the homozygous recessive genotype. Constant gene frequencies over several generations would imply that evolution was *not* taking place. Changing gene frequencies would indicate that evolution was taking place.

The conditions necessary for gene frequencies to remain constant are the following:

1. Mating must be completely random.
2. Mutations must not occur.
3. The migration of individual organisms into and out of the population must not occur.
4. The population must be very large.
5. All genes must have an equal chance of being passed on to the next generation. (Natural selection is not occurring.)

The **Hardy-Weinberg concept** states that gene frequencies will remain constant if these five conditions are met. The concept is important, because it allows a simple comparison of allele

| | | Possible female gametes | |
|---|---|---|---|
| | | $A = 0.6$ | $a = 0.4$ |
| **Possible male gametes** | $A = 0.6$ | Genotype of offspring<br>$AA = 0.6 \times 0.6 = 0.36 = 36\%$ | Genotype of offspring<br>$Aa = 0.6 \times 0.4 = 0.24 = 24\%$ |
| | $a = 0.4$ | Genotype of offspring<br>$Aa = 0.4 \times 0.6 = 0.24 = 24\%$ | Genotype of offspring<br>$aa = 0.4 \times 0.4 = 0.16 = 16\%$ |

frequency to indicate if genetic changes are occurring within a population. Two different populations of the same species can be compared to see if they have the same allele frequencies, or populations can be examined at different times to see if allele frequencies are changing.

## Determining Genotype Frequencies

It is possible to apply the Punnett square method from chapter 10 to an entire gene pool to illustrate how the Hardy-Weinberg concept works. Consider a gene pool composed of only 2 alleles, *A* and *a*. Of the alleles in the population, 60% (0.6) are *A* and 40% (0.4) are *a*. In this hypothetical gene pool, we do not know which individuals are male or female and we do not know their genotypes. With these allele frequencies, how many of the individuals would be homozygous dominant (*AA*), homozygous recessive (*aa*), and heterozygous (*Aa*)? To find the answer, we treat these alleles and their frequencies as if they were individual alleles being distributed into sperm and eggs. The sperm produced by the males of the population will be 60% (0.6) *A* and 40% (0.4) *a*. The females will produce eggs with the same relative frequencies. We can now set up a Punnett square as shown at the top of this page. The Punnett square gives the frequency of occurrence of the three possible genotypes in this population: $AA = 36\%$, $Aa = 48\%$, and $aa = 16\%$.

If we use the relationship $p^2 + 2pq + q^2 = 1$, $p^2$ is the frequency of the *AA* genotype, $2pq$ is the frequency of the *Aa* genotype, and $q^2$ is the frequency of the *aa* genotype. Then, $p^2 = 0.36$ and $p$ would be the square root of 0.36, which is 0.6—our original frequency for the *A* allele. Similarly, $q^2 = 0.16$ and $q$ would be the square root of 0.16, which is 0.4. In addition, $2pq$ would equal $2 \times 0.6 \times 0.4 = 0.48$. If this population were to reproduce randomly, it would maintain an allele frequency of 60% *A* and 40% *a* alleles. It is important to understand that Hardy-Weinberg conditions rarely exist; therefore, there are usually changes in gene frequency over time or genetic differences in separate populations of the same species. If gene frequencies are changing, evolution is taking place.

## Why Hardy-Weinberg Conditions Rarely Exist

*Random mating does not occur* for a variety of reasons. Many species are divided into small local populations that are isolated from one another and mating with individuals in other local populations rarely occurs. In human populations, these isolations may be geographic, political, or social. In addition, some individuals may be chosen as mates more frequently than others because of the characteristics they display. Therefore, the Hardy-Weinberg conditions are seldom met, because non-random mating is a factor that leads to changing gene frequencies.

**Migrating Dandelion Seeds**

*Spontaneous mutations occur.* Totally new kinds of alleles are introduced into a population, or 1 allele is converted into another, currently existing allele. Whenever an allele is changed, 1 allele is subtracted from the population and a different allele is added, thus changing the allele frequency in the gene pool. Mutations in disease-causing organisms may have significant impacts (Outlooks 13.3).

*Immigration and emigration of individual organisms are common.* When organisms move from one population to another, they carry their genes with them. Their genes are subtracted from the population they left and added to the population they enter, thus changing the gene pool of both populations. It is important to understand that migration is common for plants as well as animals. In many parts of the world, severe weather disturbances have lifted animals and plants (or their seeds) and moved them over great distances, isolating them from their original gene pool. In other instances, organisms have been distributed by floating on debris on the surface of the ocean. As an example of how important immigration and emigration is, consider the tiny island of Surtsey (3 $km^2$), which emerged from the sea as a volcano near Iceland in 1963 and continued to erupt until 1967. The new island was declared a nature preserve and has been surveyed regularly to record the kinds of organisms present. The nearest possible source of new organisms is about 20 kilometers away. The first living thing observed on the island was a fly seen less than a year after the initial eruption. By 1965, the first flowering plant had been found and, by 1996, 50 species of flowering plants had been recorded on the island. In addition, several kinds of sea birds nest on the island.

*Populations are not infinitely large,* as assumed by the Hardy-Weinberg concept. If numbers are small, random events to a few organisms might alter gene frequencies from what was expected. Consider coin flipping as an analogy. Coins have two surfaces, so, if you flip a coin once, there is a

## OUTLOOKS 13.3

# The Reemerging of Infectious Diseases

Infectious diseases caused by bacteria, viruses, fungi, and parasitic worms continue to be a major cause of suffering and death throughout the world. They are the third leading cause of death in the United States. *Reemerging infectious diseases* (for example, diphtheria, malaria, whooping cough) are diseases that were once major health concerns but then declined significantly. However, they are beginning to increase in frequency.

The reemergence of many kinds of infectious diseases is the result of two primary factors: our failure to immunize against these diseases, and evolutionary changes in the microbes. People who are not being immunized against diseases are susceptible and may become ill with the disease or become asymptomatic carriers of the microbe. A further contributing factor to the reemergence of old diseases is the increased number of people with poorly functioning immune systems. HIV/AIDS has created a huge population of people with compromised immune systems. Famine and malnutrition also impair the immune system. War and the crowding that occurs in refugee camps and prisons enhance the easy spread of disease.

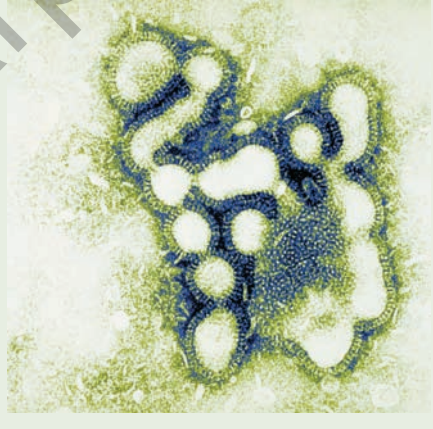
The reemergence of some diseases is also the result of evolution. Mutations are necessary if evolution is to take place. As parasites and their hosts interact, they constantly react to each other in an evolutionary fashion. Hosts develop new mechanisms to combat parasites, and parasites develop new mechanisms to overcome the hosts' defenses (for example, antibiotic resistance).

One of the mechanisms viruses use is a high rate of mutation. This ability to mutate has resulted in many new, serious human diseases. In addition, many new diseases arise when viruses that cause disease in another animal are able to establish themselves in humans. Many kinds of influenza originated in pigs, ducks, or chickens and were passed to humans through close contact with infected animals or by eating infected animals. In many parts of the world, these domesticated animals live in close contact with humans (often in the same building) making conditions favorable for the transmission of animal viruses to humans.

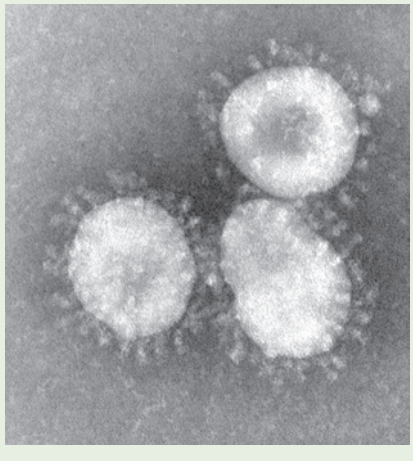
Each year, mutations result in new varieties of influenza and colds, which pass through the human population. Occasionally, the new varieties are deadly. In 1918, a new variety of influenza virus originated in pigs in the United States and spread throughout the world. During the 1918–1919 influenza pandemic that followed, 20 to 40 million people died. In 1997 in Hong Kong, a new kind of influenza was identified that killed 6 of the 18 people infected. When public health officials discovered the virus had come from chickens, they ordered the slaughter of all the live chickens in Hong Kong, which stopped the spread of the disease.

In early 2003, an outbreak of a new viral disease, known as *severe acute respiratory syndrome (SARS),* originated in China. SARS is a variation of a coronavirus, a class of virus commonly associated with the common cold, but it causes severe symptoms and, if untreated, can result in death. In June 2003, the SARS virus was isolated from an animal known as the masked palm civet (*Paguma larvata*). This animal is used for food in China and is a possible source of the virus that caused SARS in humans. However, other animals have also tested positive for the virus. The disease spread rapidly to several countries as people traveled by airplane from China to other parts of the world. A recognition of the seriousness of the disease and the isolation of infected persons prevented further spread, and this new disease was brought under control. However, if the virus still exists in some unknown wild animal host, it could reappear in the future.

In addition to cold and influenza, other kinds of diseases often make the leap from nonhuman to human hosts. The swine flu virus outbreaks of 2009 were traced to a population of pigs in Mexico. It is likely that genetic mixing occurred in the pigs' cells, resulting in a new kind of virus containing bird, human, and swine genes. The emergence of these new kinds of viruses enables them to more easily move from one species to another.



*(a)* Influenza Viruses at 295,000 Magnification



*(b)* Three SARS Virus Particles