chapter 13 Genetics and Biotechnology

section 3 The Human Genome

MAIN Idea

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

What You'll Learn

- how forensic scientists use DNA fingerprinting
- how human genome information can help diagnose diseases

Before You Read

Scientists now study genes in ways that were not invented 20 years ago. Think of the new technology in your own life. What are some new technologies you use?

_____

_____

Study Coach

Make Flash Cards Make a flash card for each key term in this section. Write the term on one side of the card. Write the definition on the other side. Use the flash cards to review what you have learned.

Read to Learn

The Human Genome Project

A genome is all of the genetic information in a cell. The human genome is all of the genetic information in a human cell. The Human Genome Project (HGP) was an enormous project. One goal was to learn the sequence of the billions of nucleotides that make up human DNA. Another goal was to identify all 20,000 to 25,000 human genes.

The HGP was completed in 2003. Scientists will be working for many years to understand the data.

How was the human genome sequenced?

Human DNA is organized into 46 chromosomes. To determine the human genome, each chromosome was cut. Several restriction enzymes were used to make fragments with overlapping sequences. The fragments were combined with vectors and copied. The overlapping sequences were analyzed to generate a continuous sequence.

As scientists studied the sequences in the human genome, they observed that less than 2 percent of all of the nucleotides in the human genome code for all of the proteins in the body. The rest of the DNA is made of long stretches of repeated sequences called noncoding sequences. Scientists do not yet know the function of these sequences.

Applying Math

1. **Calculate** What percentage of human DNA is not made of genes?

_____

How is DNA fingerprinting used?

The protein-coding sections of DNA are almost identical from one person to the next. The long stretches of noncoding sections of DNA are unique to each individual. **DNA fingerprinting** uses gel electrophoresis to observe the patterns that are unique to each person.

Forensic scientists use DNA fingerprinting to identify suspects and victims in a crime. DNA fingerprinting has been used to convict criminals and free innocent people who were wrongly imprisoned. DNA fingerprinting can be used to identify soldiers killed in war and establish paternity.

When only a drop of blood or a single hair is found at a crime scene, the sample does not contain enough DNA for DNA fingerprinting. Forensic scientists use PCR to copy the DNA and make a larger sample. The DNA is then cut with restriction enzymes and separated by gel electrophoresis. The pattern of the fragments from the sample is compared with DNA samples from known sources, such as a suspect or a victim in a crime.

Identifying Genes

Once the genome has been sequenced, the next step is to identify the genes and determine their functions. Organisms, such as bacteria and yeast, do not have noncoding DNA. Scientists look for DNA sequences called open reading frames (ORFs). ORFs are made of codons—groups of three nucleotides that code for amino acids. ORFs begin with a start codon and end with a stop codon. In between the start and stop codons, ORFs contain at least 100 codons. Scientists have identified over 90 percent of genes in yeast and bacteria by looking for ORFs.

In humans and other complex organisms, the long stretches of noncoding sequence make looking for genes more difficult. Scientists use sophisticated computer programs called algorithms to identify genes.

Bioinformatics

The sequencing of DNA from humans and other organisms has created large amounts of data. It has also led to a new field of study. **Bioinformatics** is the study of how to create and use computer databases to store, organize, index, and analyze this data. Scientists are using bioinformatics to discover new ways to locate genes in DNA sequences and to study the evolution of genes.

Think it Over

2. Identify What is most useful for DNA fingerprinting: protein-coding sequences or noncoding sequences? Explain.

Reading Check

3. Explain What is bioinformatics?

DNA Microarrays

In any cell at any time, some genes are expressed, meaning those genes are making proteins. The rest of the genes are silent. In a different cell or at a different time, other genes will be expressed.

Reading Check

4. Define What does it mean to say a gene is expressed?

_____

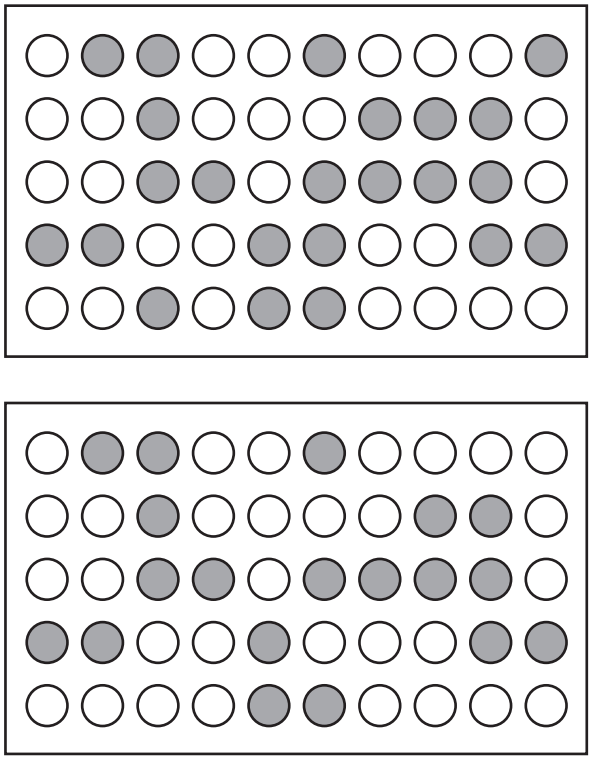
_____

DNA microarrays are tiny microscope slides or silicon chips that contain tiny spots of DNA fragments. One microarray can contain thousands of genes. Scientists use DNA microarrays to study the expression of a lot of genes at once. DNA microarrays are used to study when and where genes are expressed. Microarrays can reveal how gene expression changes under different conditions. Microarrays can be used to compare cancer cells to normal cells. By finding genes that are expressed in cancer cells, scientists can learn more about cancer. They can learn better ways to treat people with cancer.

The figure below shows two DNA microarrays. Each spot represents a different gene. Spots that are white indicate the gene is being expressed. Spots that are black indicate the gene is not being expressed. The top microarray shows the genes that are expressed in a normal cell. The bottom microarray shows the genes that are expressed in a cancer cell.

Picture This

5. Analyze Find the genes that are expressed in the cancer cell but not in the normal cell. Circle the spots that represent those genes.



The Genome and Genetic Disorders

Over 99 percent of all nucleotide sequences are exactly the same from one person to the next. **Single nucleotide polymorphisms**, or SNPs, are variations in the DNA sequence that occur when a single nucleotide in the genome is changed. A variation is only considered an SNP if it occurs in at least 1 percent of the population.

SNPs can be useful to scientists. Many SNPs do not change how cells function, but SNPs might help scientists find other, nearby mutations that do cause genetic disease. Some SNPs occur near mutations that cause human diseases. Knowing where SNPs occur in the genome might help scientists find mutations that cause diseases.

What is the HapMap project?

A group of international scientists is creating a list of common genetic variations in people. Genetic variations located close together on a chromosome are said to be linked. Linked variations are usually inherited together.

A **haplotype** is a section of linked variations in the human genome. The haplotype map or HapMap project is an international effort to find all the haplotypes. The project will describe what these variations are and show where they are found. The HapMap project will also describe how these variations occur among people within populations and among populations from different areas of the world.

The HapMap project will enable scientists to take advantage of how SNPs and other genetic variations are organized on chromosomes. This will help scientists find genes that cause different types of disease. The HapMap will also help scientists find mutations that affect how a person responds to medicine.

What is pharmacogenomics?

One day people might go to the doctor and have drugs specially prescribed for them based on their genes. **Pharmacogenomics** (far muh koh jeh NAW mihks) is the study of how a person's genes affect his or her response to medicine.

Researchers hope that pharmacogenomics will allow drugs to be custom made for people based on their genetic makeup. Pharmacogenomics might allow doctors to prescribe drugs that are safer, more specific, more effective, and have fewer side effects. Doctors might one day read your genetic code and prescribe drugs made especially for you.

Applying Math

6. Calculate A single nucleotide variation occurs in 7 of every 1000 people. Is this variation an SNP? Why or why not?

Reading Check

7. Define What is a possible benefit of pharmacogenomics?

How does gene therapy work?

Gene therapy is a way of fixing mutated genes that cause disease. Scientists insert a normal gene into a chromosome to replace the mutated gene. The normal gene can then do the work of the mutated gene.

A virus is used as a vector to transfer the normal gene to the cell. The virus releases the recombinant DNA, which contains the normal gene, into the cell. The normal gene inserts itself into the genome and begins functioning.

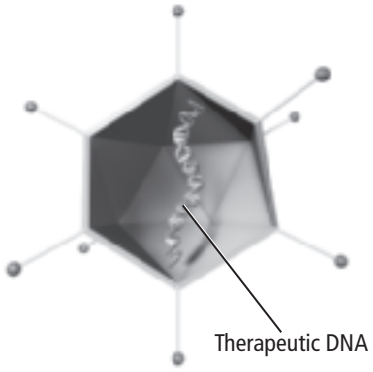
The Food and Drug Administration monitors new medical trials, including gene therapy. Although there have been setbacks, recent trials include work with diabetes, cancer, and retinal disease.

Picture This

8. Identify How is a normal gene inserted into a cell? (Circle your answer.)

a. by a virus releasing recombinant DNA containing the normal gene

b. by physically removing the mutated gene



Genomics and Proteomics

Genomics is the study of an organism's genome. Following the completion of the human genome sequence in 2003, so much research has become focused on genomics that biologists call this "the genomic era."

Genomics is a powerful strategy for identifying human genes and understanding how they work. Researchers also use genomics to study plants and other organisms, such as rice, mice, fruit flies, and corn, whose genomes have been sequenced.

Genes are important because they are the way cells store information. Proteins are important because they are the machines that make cells run.

Proteomics is the large-scale study and cataloging of the structure and function of proteins in the human body. With proteomics, researchers can study hundreds or thousands of proteins at one time.

Scientists use proteomics to understand human diseases. Scientists expect that proteomics will change the development of medicines to treat diseases such as diabetes, obesity, and atherosclerosis.

Reading Check

9. Define What is proteomics?