

Reading Preview

Key Concepts

- What are two major causes of genetic disorders in humans?
- How do geneticists trace the inheritance of traits?
- How are genetic disorders diagnosed and treated?

Key Terms

- genetic disorder
- pedigree
- karyotype



Target Reading Skill

Comparing and Contrasting

As you read, compare and contrast the types of genetic disorders by completing a table like the one below.

Disorder	Description	Cause
Cystic fibrosis	Abnormally thick mucus	Loss of three DNA bases

Lab Zone

Discover Activity

How Many Chromosomes?

The photo at right shows the chromosomes from a cell of a person with Down syndrome, a genetic disorder. The chromosomes have been sorted into pairs.

1. Count the number of chromosomes in the photo.
2. How does the number of chromosomes compare to the usual number of chromosomes in human cells?



Think It Over

Inferring How do you think a cell could have ended up with this number of chromosomes? (*Hint:* Think about the events that occur during meiosis.)

The air inside the stadium was hot and still. The crowd cheered loudly as the runners approached the starting blocks. At the crack of the starter's gun, the runners leaped into motion and sprinted down the track. Seconds later, the race was over. The runners, bursting with pride, hugged each other and their coaches. These athletes were running in the Special Olympics, a competition for people with disabilities. Many of the athletes who compete in the Special Olympics have disabilities that result from genetic disorders.



Runners in the Special Olympics



FIGURE 9
Sickle-Cell Disease
 Normally, red blood cells are shaped like round disks (top). In a person with sickle-cell disease, red blood cells can become sickle-shaped (bottom).

Lab
zone

Skills Activity

Predicting

A man has sickle-cell disease. His wife does not have the disease, but is heterozygous for the sickle-cell trait. Predict the probability that their child will have sickle-cell disease. (Hint: Construct a Punnett square.)

Causes of Genetic Disorders

A **genetic disorder** is an abnormal condition that a person inherits through genes or chromosomes. Some genetic disorders are caused by mutations in the DNA of genes. Other disorders are caused by changes in the overall structure or number of chromosomes. In this section, you will learn about some common genetic disorders.

Cystic Fibrosis Cystic fibrosis is a genetic disorder in which the body produces abnormally thick mucus in the lungs and intestines. The thick mucus fills the lungs, making it hard for the affected person to breathe. Cystic fibrosis is caused by a recessive allele on one chromosome. The recessive allele is the result of a mutation in which three bases are removed from a DNA molecule.

Sickle-Cell Disease Sickle-cell disease affects hemoglobin, a protein in red blood cells that carries oxygen. When oxygen concentrations are low, the red blood cells of people with the disease have an unusual sickle shape. Sickle-shaped red blood cells clog blood vessels and cannot carry as much oxygen as normal cells. The allele for the sickle-cell trait is codominant with the normal allele. A person with two sickle-cell alleles will have the disease. A person with one sickle-cell allele will produce both normal hemoglobin and abnormal hemoglobin. This person usually will not have symptoms of the disease.

Hemophilia Hemophilia is a genetic disorder in which a person's blood clots very slowly or not at all. People with the disorder do not produce one of the proteins needed for normal blood clotting. The danger of internal bleeding from small bumps and bruises is very high. Hemophilia is caused by a recessive allele on the X chromosome. Because hemophilia is a sex-linked disorder, it occurs more frequently in males than in females.

Down Syndrome In Down syndrome, a person's cells have an extra copy of chromosome 21. In other words, instead of a pair of chromosomes, a person with Down syndrome has three of that chromosome. Down syndrome most often occurs when chromosomes fail to separate properly during meiosis. People with Down syndrome have some degree of mental retardation. Heart defects are also common, but can be treated.



Reading
Checkpoint

How is the DNA in the sickle-cell allele different from the normal allele?

Pedigrees

Imagine that you are a geneticist who is interested in tracing the occurrence of a genetic disorder through several generations of a family. What would you do? **One important tool that geneticists use to trace the inheritance of traits in humans is a pedigree.** A pedigree is a chart or "family tree" that tracks which members of a family have a particular trait.

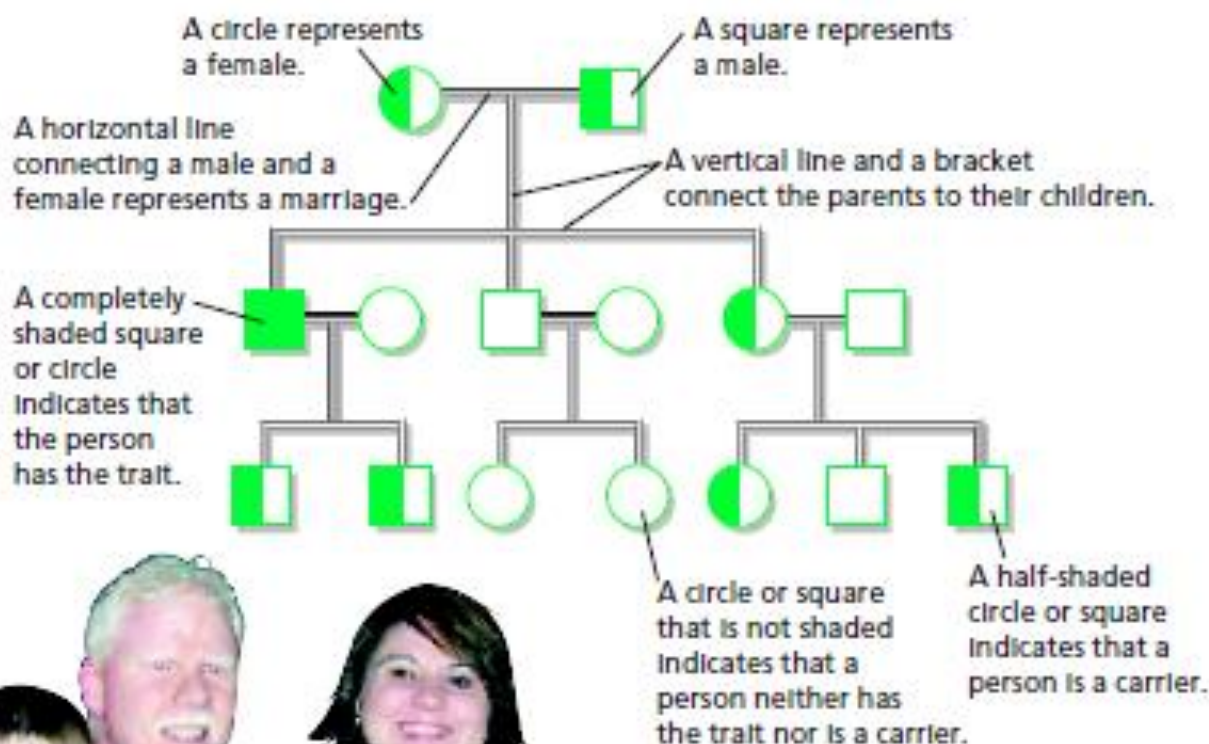
The trait in a pedigree can be an ordinary trait, such as a widow's peak, or a genetic disorder, such as cystic fibrosis. Figure 10 shows a pedigree for albinism, a condition in which a person's skin, hair, and eyes lack normal coloring.

FIGURE 10
A Pedigree

The father in the photograph has albinism. The pedigree shows the inheritance of the allele for albinism in three generations of a family. **Interpreting Diagrams** Where is an albino male shown in the pedigree?

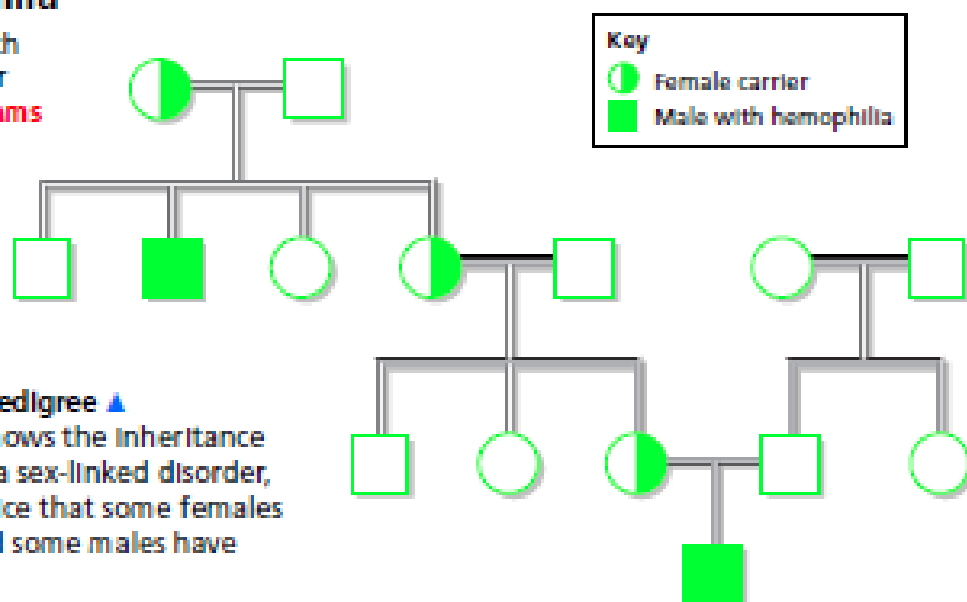
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Living With Hemophilia

With proper care, people with hemophilia can manage their disorder. **Interpreting Diagrams** In the pedigree, how many people have hemophilia?



A Hemophilia Pedigree

The pedigree shows the inheritance of hemophilia, a sex-linked disorder, in a family. Notice that some females are carriers, and some males have the disorder.

Managing Genetic Disorders

Years ago, doctors had only Punnett squares and pedigrees to help them predict whether a child might have a genetic disorder. Today, doctors use tools such as karyotypes to help diagnose genetic disorders. People with genetic disorders are helped through medical care, education, job training, and other methods.

Karyotypes To detect chromosomal disorders such as Down syndrome, a doctor examines the chromosomes from a person's cells. The doctor uses a karyotype to examine the chromosomes. A **karyotype** (KA ree uh typ) is a picture of all the chromosomes in a cell. The chromosomes in a karyotype are arranged in pairs. A karyotype can reveal whether a person has the correct number of chromosomes in his or her cells. If you did the Discover activity, you saw a karyotype from a girl with Down syndrome.

Genetic Counseling A couple that has a family history of a genetic disorder may turn to a genetic counselor for advice. Genetic counselors help couples understand their chances of having a child with a particular genetic disorder. Genetic counselors use tools such as karyotypes, pedigree charts, and Punnett squares to help them in their work.



What do genetic counselors do?

Physical Therapy ▶

Trained medical workers help hemophilia patients cope with their disorder. Here, a boy receives physical therapy.



Sports ▶

A boy with hemophilia learns how to play golf. The disorder does not stop people from living active lives.



Dealing With Genetic Disorders People with genetic disorders face serious challenges, but help is available. Medical treatments help people with some disorders. For example, physical therapy helps remove mucus from the lungs of people with cystic fibrosis. People with sickle-cell disease take folic acid, a vitamin, to help their bodies manufacture red blood cells. Because of education and job training, adults with Down syndrome can find work in hotels, banks, restaurants, and other places of employment. Fortunately, most genetic disorders do not prevent people from living active, productive lives.

Section 2 Assessment

Target Reading Skill

Comparing and Contrasting Use the information in your table to help you answer Question 1 below.

Reviewing Key Concepts

- Identifying** Identify the two major causes of genetic disorders in humans.
 - Explaining** Which of those two major causes is responsible for Down syndrome?
 - Describing** How are the cells of a person with Down syndrome different from those of a person without the disorder?
- Defining** What is a pedigree?
 - Inferring** Why are pedigrees helpful in understanding genetic disorders?

- Applying Concepts** Sam has hemophilia. Sam's brother, mother, and father do not have hemophilia. Draw a pedigree showing who has the disorder and who is a carrier.
- Reviewing** What is a karyotype?
 - Inferring** Would a karyotype reveal the presence of sickle-cell disease? Why or why not?

Writing in Science

Creating a Web Site Create an imaginary Web site to inform the public about genetic disorders. Write a description of one disorder for the Web site.