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Human Genetic Disorders (pages 117-121)

Causes of Genetic Disorders (page 118)

Key Concept: 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.

- A genetic disorder is an abnormal condition that a person inherits through genes or chromosomes.
- Some genetic disorders are caused by a change in DNA. Cystic fibrosis is caused by a mutation in a gene.
 People with cystic fibrosis have abnormally thick mucus in their lungs. The allele for cystic fibrosis is recessive to the normal allele.
- Other genetic disorders are caused by changes in chromosome number or structure. In Down syndrome, a person has one extra copy of chromosome 21. People with Down syndrome may have some mental retardation as well as heart problems.

Answer the following questions. Use your textbook and the ideas above.

- 1. Circle the letter of each cause of a genetic disorder.
 - a. changes in the environment
 - b. changes in DNA
 - c. changes in chromosome number or structure
- 2. Circle the letter of each genetic disorder.
 - a. Down syndrome
 - b. chicken pox
 - c. cystic fibrosis

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Pedigrees (page 119)

Key Concept: 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 the members of a family that have a certain trait.
- Geneticists use pedigrees to follow a human trait through several generations of a family. This helps geneticists learn how the trait is inherited.
- In a pedigree, a circle stands for a female (woman). A square stands for a male (man). A line connecting a square and circle shows that the man and woman are married. Children are connected to their parents by a line and a bracket.
- A shaded square or circle stands for a person who has
 the trait. A half-shaded square or circle stands for a
 person who carries just one allele for the trait, but does
 not show the trait. A person who carries one allele for a
 trait, but does not have the trait is called a carrier. A
 circle or square that is not shaded stands for a person
 who does not have the trait.

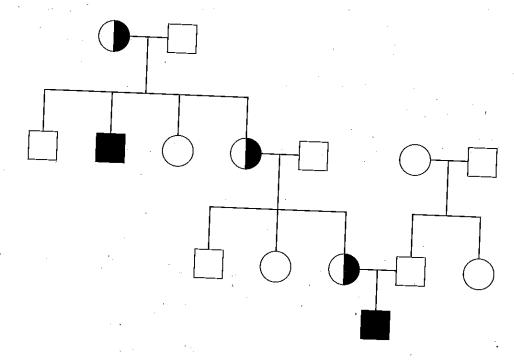
Answer the following questions. Use your textbook and the ideas above.

- **3.** What is a chart that follows a trait through several generations of a family? Circle the letter of the correct answer.
 - a. Punnett square
 - b. allele combinations
 - c. pedigree

4. Look at the symbol below.



- Does this symbol stand for a male or a female?
- b. Does this symbol stand for a person who has the trait or one who is a carrier for the trait?
- 5. The chart below follows hemophilia in a family. Hemophilia is a genetic disorder in which the blood does not clot normally. How many males (men) have hemophilia? Circle the letter of the correct answer.
 - 1 a.
 - **b.** 2
 - **c.** 3



Managing Genetic Disorders

(pages 120-121)

Key Concept: 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.

- A karyotype is a picture of all the chromosomes in a cell. A karyotype shows whether or not a person has the correct number of chromosomes.
- Genetic counselors help couples understand their chances of having a child with a genetic disorder.
 Genetic counselors use karyotypes, pedigree charts, and Punnett squares in their work.
- People with genetic disorders live active, productive lives. Some people might need medical care. Others might need education and job training.

Answer the following questions. Use your textbook and the ideas above.

6. Read each word in the box. In each sentence below, fill in the correct word or words.

genetic counselor karyotype pedigree

- a. A picture of all the chromosomes in a cell is a
- **b.** A person who explains the chances of having a child with a genetic disorder is a
- 7. Is the following sentence true or false? It is not possible to help people who have genetic disorders. _____