Name _____ Date _____ Class _____

SECTION 4-2 SECTION SUMMARY

Human Genetic Disorders

Guide for Reading

- **What causes genetic disorders?**
- **How are genetic disorders diagnosed?**

A **genetic disorder** is an abnormal condition that a person inherits through genes or chromosomes. **Genetic disorders are caused by mutations, or changes in a person's DNA.**

Cystic fibrosis is a genetic disorder in which the body produces abnormally thick mucus in the lungs and intestines, making it hard to breathe and digest food. The allele that causes cystic fibrosis is recessive. Currently there is no cure for cystic fibrosis, although there are treatments to help control the symptoms.

Sickle-cell disease is a genetic disorder caused by abnormal hemoglobin, the protein in blood that carries oxygen. People with sickle-cell disease suffer from lack of oxygen in the blood and experience pain and weakness. The allele that causes sickle-cell disease is codominant with the normal allele. People with two sickle-cell alleles have the disease. People with one sickle-cell allele produce both normal and abnormal hemoglobin but usually do not have symptoms of the disease. Currently there is no cure for sickle-cell disease. However, treatments can lessen the pain and other symptoms.

Hemophilia is a genetic disorder in which the blood clots very slowly or not at all. People with the disorder do not produce one of the proteins needed for normal blood clotting. Hemophilia is caused by a recessive allele on the X chromosome. Because it is a sex-linked disorder, it occurs more often in males than in females. With treatment, people with hemophilia can lead normal lives.

Down syndrome is a genetic disorder that is due to an extra copy of chromosome 21. Most often this occurs when the chromosomes fail to separate properly during meiosis. People with Down syndrome have a distinctive physical appearance and some degree of mental retardation. Many people with Down syndrome lead full, active lives.

Today doctors use tools such as amniocentesis and karyotypes to help detect genetic disorders. Before a baby is born, doctors can use a procedure called **amniocentesis** to determine whether the baby will have some genetic disorders. The chromosomes in a small amount of fluid surrounding the developing baby are examined. The doctor creates a **karyotype,** or picture of all the chromosomes in a cell, arranged in pairs. A karyotype can reveal whether a developing baby has the correct number of chromosomes in its cells and whether it is a boy or a girl.

A couple that has a family history or concern about 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.

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SECTION 4-2 REVIEW AND REINFORCE

Human Genetic Disorders

◆ Understanding Main Ideas

Complete the table below. Then answer the questions that follow.

Human Genetic Disorders

Disorder	Type of Allele	Effects on Body
1.	**2.**	Abnormally thick mucus in lungs and intestines
Sickle-cell disease	**3.**	**4.**
5.	Recessive sex-linked	Blood clots poorly

Write your answers on a separate sheet of paper.

6. Name one treatment for cystic fibrosis.

7. How does sickle-cell trait differ from sickle-cell disease?

8. Why is hemophilia more common in males than in females?

9. Explain what causes Down syndrome.

◆ Building Vocabulary

If the statement is true, write true. If the statement is false, change the underlined word or words to make the statement true.

_____ **10.** A genetic disorder is an abnormal condition that a person inherits through genes or chromosomes.

_____ **11.** Genetic counseling is a procedure in which cells are examined to determine whether a baby will have some genetic disorders.

_____ **12.** A picture of all the chromosomes in a cell is called a genotype.