Modern Genetics • Guided Reading and Study

Human Genetic Disorders

This section describes how changes in the DNA of some genes have affected certain traits in humans.

Use Target Reading Skills

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

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Description</th>
<th>Cause</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cystic fibrosis</td>
<td>Abnormally thick mucus</td>
<td>Loss of three DNA bases</td>
</tr>
<tr>
<td>Sickle-cell disease</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hemophilia</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Down syndrome</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Causes of Genetic Disorders

1. An abnormal condition that a person inherits through genes or chromosomes is called a(n) _________________.

2. What causes genetic disorders?

______________________________

______________________________

______________________________
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3. What is cystic fibrosis?

4. Is the following sentence true or false? Cystic fibrosis is caused by a mutation that is the dominant allele of a gene. ____________________

5. Circle the letter of the protein that is not normal in people with sickle-cell disease.
   a. mucus
   b. hemoglobin
   c. red blood cells
   d. clotting protein

6. The allele for the sickle-cell trait is ____________________ with the normal allele.

7. Is the following sentence true or false? Hemophilia is caused by a dominant allele on the X chromosome. ____________________

8. Hemophilia occurs more often in ____________________.

   a. recessive allele
   b. dominant allele
   c. too many chromosomes
   d. too few chromosomes

10. Down syndrome most often occurs when ____________________ fail to separate properly during meiosis.

Pedigrees

11. A chart or “family tree” that tracks which members of a family have a certain trait is called a(n) ____________________.

12. Is the following sentence true or false? On a pedigree, a circle represents a male. ____________________

Managing Genetic Disorders

13. How are people helped when they have a genetic disorder?

14. A ____________________ is a picture of all the chromosomes in a cell.
Modern Genetics • Section Summary

Human Genetic Disorders

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?

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.

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 that affects 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 Down syndrome 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.

Geneticists trace the inheritance of traits through several generations of a family. 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 have a particular trait.

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. To detect chromosomal disorders such as Down syndrome, a doctor examines the chromosomes from a person’s cells. The doctor uses a karyotype, or picture of all the chromosomes in a cell, to examine the chromosomes. The chromosomes are arranged in pairs. A karyotype can reveal whether a developing baby has the correct number of chromosomes in its cells.

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.
Human Genetic Disorders

Understanding Main Ideas
Complete the table below. Then answer the questions that follow.

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Type of Allele</th>
<th>Effects on Body</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. _______________</td>
<td>2. _______________</td>
<td>Abnormally thick mucus in lungs and intestines</td>
</tr>
<tr>
<td>Sickle-cell disease</td>
<td>3. _______________</td>
<td>4. _______________</td>
</tr>
<tr>
<td>5. _______________</td>
<td>Recessive sex-linked</td>
<td>Blood clots poorly.</td>
</tr>
</tbody>
</table>

Write your answers on a separate sheet of paper.

6. How does sickle-cell trait differ from sickle-cell disease?
7. Why is hemophilia more common in males than in females?
8. Explain what causes Down syndrome.

Building Vocabulary
If the statement is true, write true. If it is false, change the underlined word to make it true.

__________ 9. A genetic disorder is an abnormal condition that a person inherits through genes or chromosomes.
__________ 10. Genetic counseling is a chart or “family tree” that tracks which members of a family have a particular trait.
__________ 11. A picture of all the chromosomes in a cell is called a genotype.
Sickle-Cell Allele and Malaria

The allele for sickle-cell disease is most common in people of African ancestry. The reason for this probably has to do with the relationship between the sickle-cell trait and malaria. Malaria, a disease common in parts of Africa, affects red blood cells. Carriers of the sickle-cell allele are resistant to malaria. Scientists think that the sickle-cell trait helps carriers resist malaria. The map on the left shows the distribution of malaria worldwide today. The map on the right shows the distribution of the sickle-cell allele.

1. Where is malaria most common today?
2. Where is the sickle-cell allele most frequent?
3. Malaria is caused by a microscopic parasite that infects the blood. Based on this fact, hypothesize why people with sickle-cell trait are resistant to malaria. (Hint: A parasite is an organism that lives and feeds on or in another organism.)
4. Suppose malaria were eliminated as a human disease. Predict how the frequency of the sickle-cell allele might change over time. Explain your prediction.