

Human Genetic Disorders

Imagine that you are reading directions to put a bicycle together. Suppose the directions say to put the handlebars where the seat should go. If you did this, your bicycle would not function the way it should. A similar thing can happen if a mutation, or change, to a gene occurs. An organism with a mutation cannot function as it should. ✓

What are the effects of genetic disorders?

A **genetic disorder** happens when a gene or chromosomal mutation is inherited. Genetic disorders can result in minor or major health problems, or even lead to death. Cystic fibrosis is a common recessive disorder among Caucasians. A recessive phenotype such as cystic fibrosis occurs when two recessive alleles for a trait are inherited.

What problems does cystic fibrosis cause?

People with cystic fibrosis have tissues that produce abnormally thick mucus. This mucus can affect the functions of the respiratory, digestive, and reproductive systems. Without treatment, children with cystic fibrosis usually live five years or less. With treatment, a child with cystic fibrosis might live into their twenties or longer. Some other human genetic disorders are listed in the table below.

Some Human Genetic Disorders		
Genetic Disorder	Type of Disorder	Major Problems
Huntington's disease	Dominant	breakdown of brain tissue; shortened life span
Sickle-cell disease	Codominant	red blood cell destruction; clogged blood vessels
Cystic fibrosis	Recessive	abnormally thick mucus; affects many organ systems
Hemophilia	X-linked recessive	excessive bleeding due to blood clotting problems
Down syndrome	Trisomy—extra chromosome #21	mental disability; heart defects

✓ Reading Check

8. Explain How does a mutation affect an organism?

Picture This

9. Highlight the genetic disorder that affects the blood.