

14.2 Human Genetic Disorders

Lesson Objectives

-  Explain how small changes in DNA cause genetic disorders.
-  Summarize the problems caused by nondisjunction.

Lesson Summary

From Molecule to Phenotype There is a molecular reason for genetic disorders. A change in DNA can alter an amino acid sequence, which can change a protein and therefore, the phenotype. Some common inherited disorders result from a change in DNA. They include:

- ▶ sickle cell disease, in which a defective polypeptide makes hemoglobin in the blood less soluble;
- ▶ cystic fibrosis, in which a deletion of three bases in a gene causes cell membranes to lose their ability to transport chloride ions;
- ▶ Huntington's disease, in which a single codon for a certain amino acid repeats more than 40 times, causing mental deterioration and uncontrolled movements.

Some alleles that cause disease in the homozygote can provide an advantage in the heterozygote. The geographic associations between sickle cell disease and malaria and between cystic fibrosis and typhoid demonstrate how the heterozygous state reduces the risk of infection.

Chromosomal Disorders Sometimes, during meiosis, homologous chromosomes fail to separate. This **nondisjunction** (not coming apart) can create a gamete with an abnormal number of chromosomes, leading to offspring with missing or extra chromosomes. Examples include:

- ▶ Down syndrome, most often a result of three copies of chromosome 21;
- ▶ Turner's syndrome, a female with a single X chromosome;
- ▶ Klinefelter's syndrome, a male with an extra X chromosome.

From Molecule to Phenotype

1. The boxes below each show a step to explain how genetic disorders have a molecular basis. Number them so that the steps are in the correct order.

A change in phenotype results. _____

A gene's DNA sequence changes. _____

The amino acid sequence that alters a protein changes. _____

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