



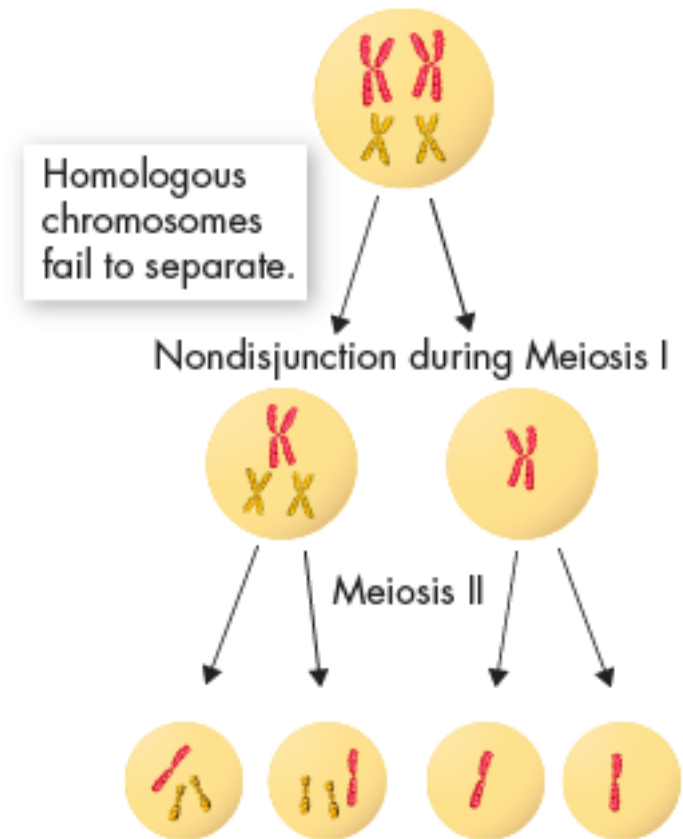
## Lesson Overview

15.2 Human  
Genetic Disorders

# Chromosomal Disorders

The most common error in meiosis occurs when homologous chromosomes fail to separate. This mistake is known as **nondisjunction**, which means “not coming apart.”

Nondisjunction may result in gametes with an abnormal number of chromosomes, which can lead to a disorder of chromosome numbers.



# Chromosomal Disorders

If two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with three copies of that chromosome.

This condition is known as a trisomy, meaning “three bodies.”

The most common form of trisomy, involving three copies of chromosome 21, is Down syndrome, which is often characterized by mild to severe mental retardation and a high frequency of certain birth defects.

# Chromosomal Disorders

Nondisjunction of the X chromosomes can lead to a disorder known as Turner's syndrome.

A female with Turner's syndrome usually inherits only one X chromosome.

Women with Turner's syndrome are sterile, which means that they are unable to reproduce. Their sex organs do not develop properly at puberty.

# Chromosomal Disorders

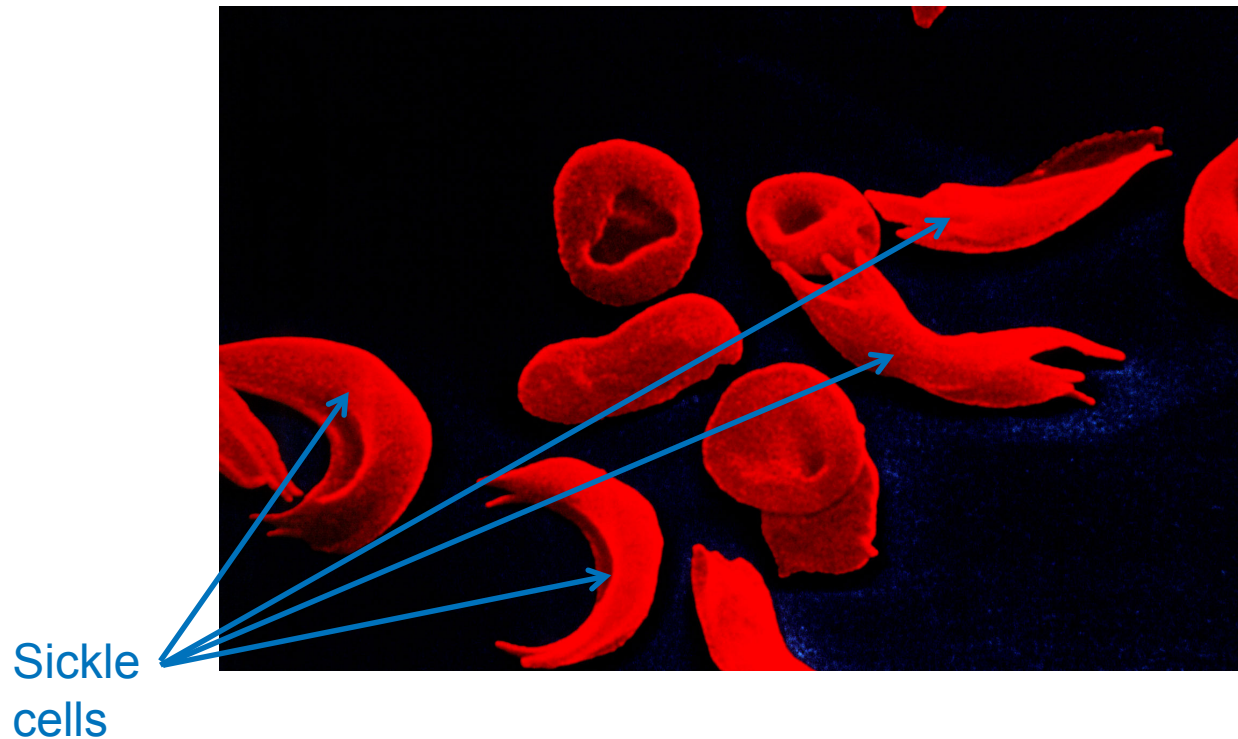
In males, nondisjunction may cause Klinefelter's syndrome, resulting from the inheritance of an extra X chromosome, which interferes with meiosis and usually prevents these individuals from reproducing.

There have been no reported instances of babies being born without an X chromosome, indicating that this chromosome contains genes that are vital for the survival and development of the embryo.

# From Molecule to Phenotype

Changes in the DNA sequence of a gene can change proteins:

- Altering amino acid sequences
- Affecting the phenotype



# Cystic Fibrosis

- Most cases result from the deletion of three bases in the DNA of a single gene
- Body does not produce normal CFTR





# Genetic Advantages

Some alleles that cause disease may also have benefits.

- Carrying one copy of the sickle cell provides protection from malaria.
- Carrying one copy of the CF allele provides protection from typhoid bacterium.

