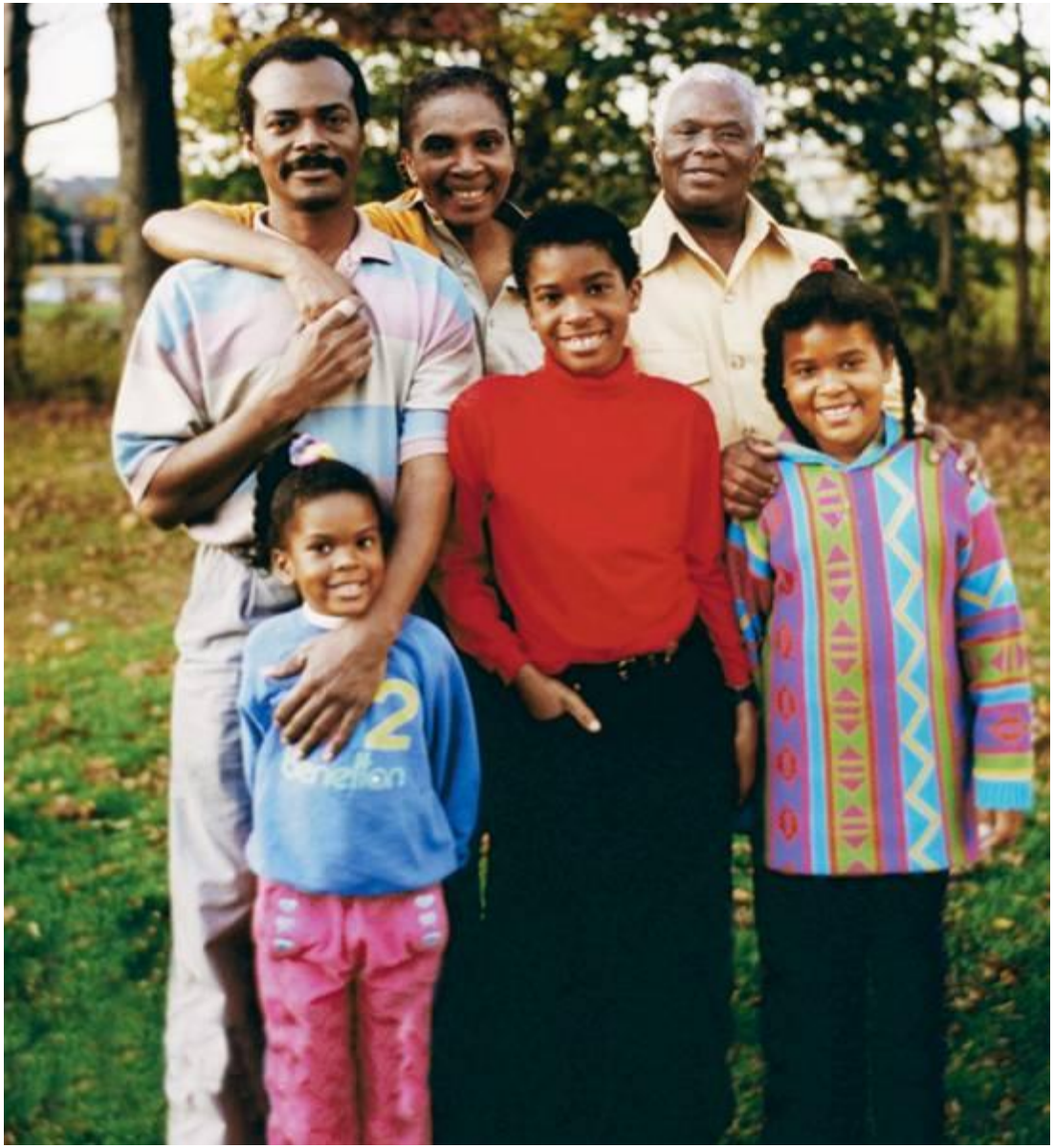
14–2 Human Chromosomes



Copyright Pearson Prentice Hall

Sex-Linked Genes

The X chromosome and the Y chromosomes determine sex.

Genes located on these chromosomes are called **sex-linked genes**.

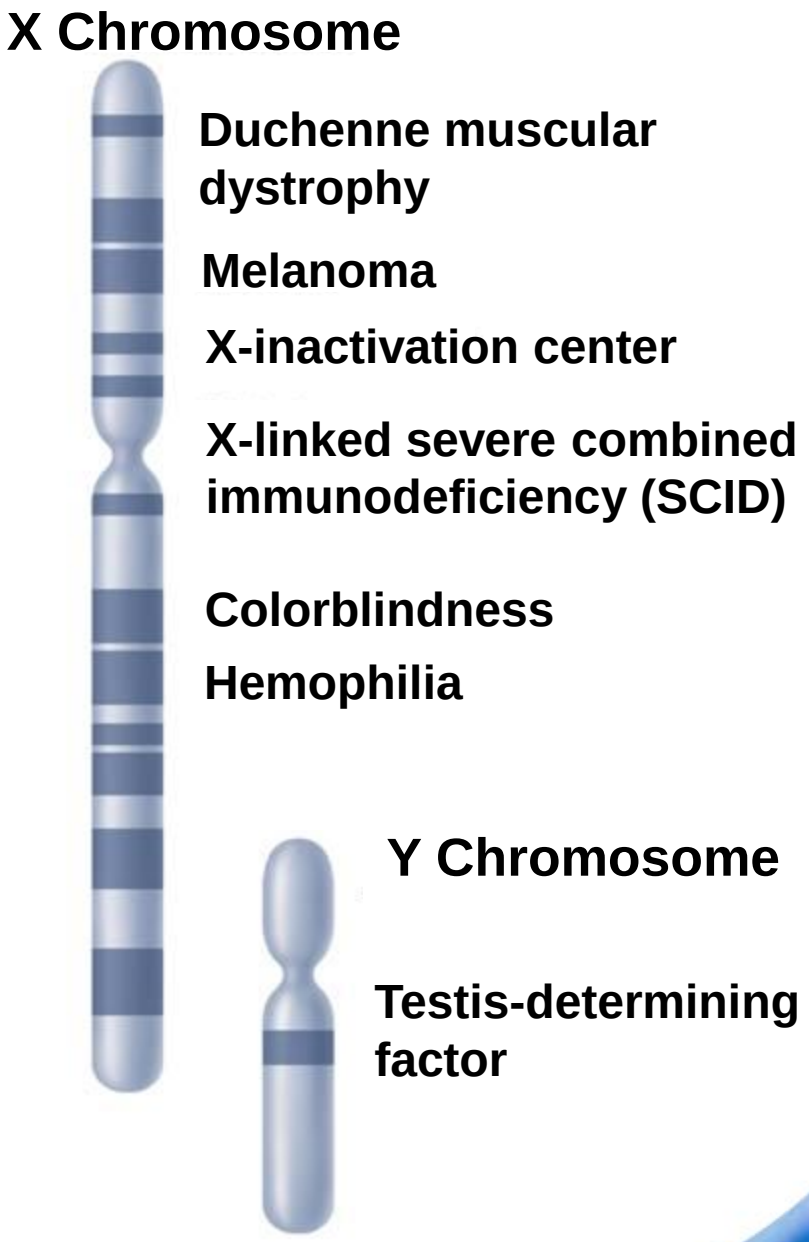
More than 100 sex-linked genetic disorders have now been mapped to the X chromosome.

An average of 3,000 base pairs exist in each gene.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

The Y chromosome is much smaller than the X chromosome and appears to contain only a few genes.



PEARSON Prentice Hall

Copyright Pearson Prentice Hall

For a recessive allele to be expressed in females, there must be two copies of the allele, one on each of the two X chromosomes.

Males have just one X chromosome. Thus, all X-linked alleles are expressed in males, even if they are recessive.

PEARSON Prentice Hall

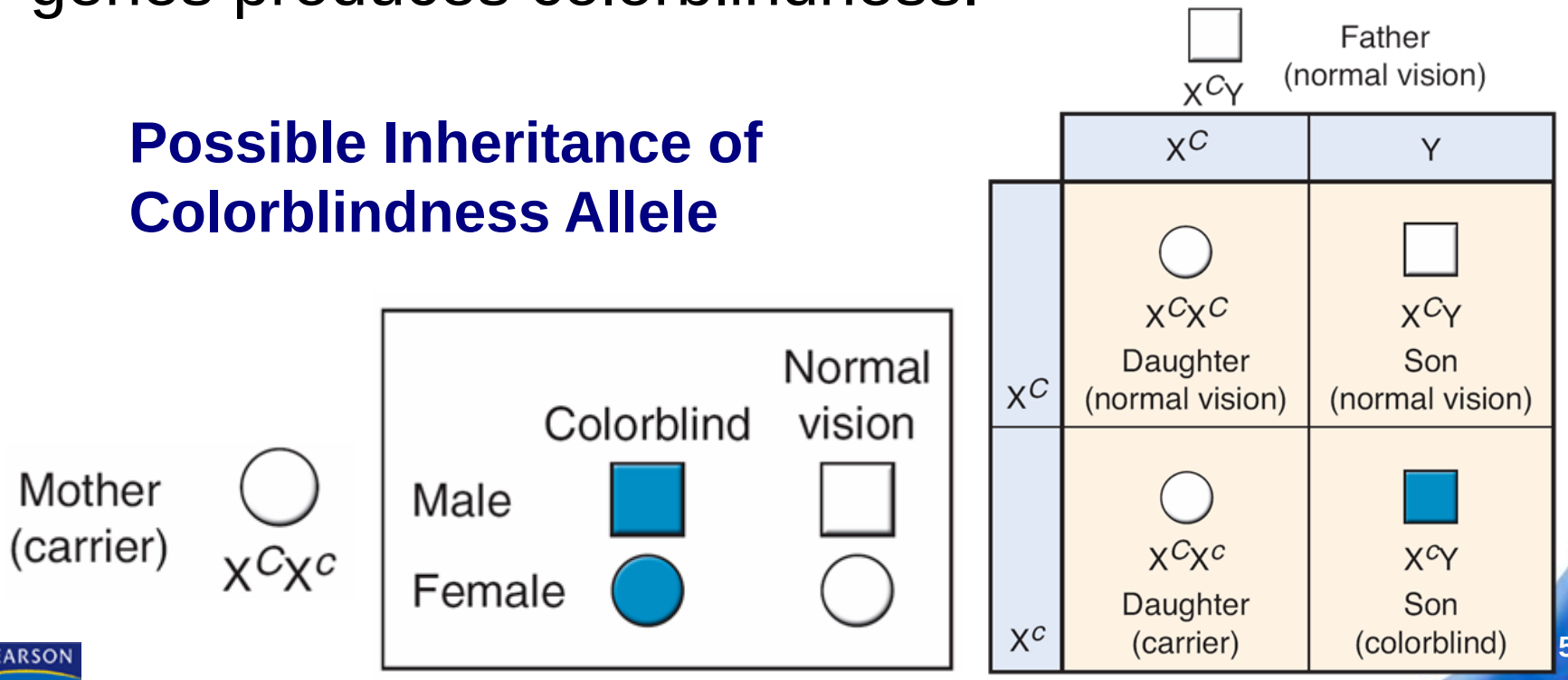
Copyright Pearson Prentice Hall

Colorblindness

Three human genes associated with color vision are located on the X chromosome.

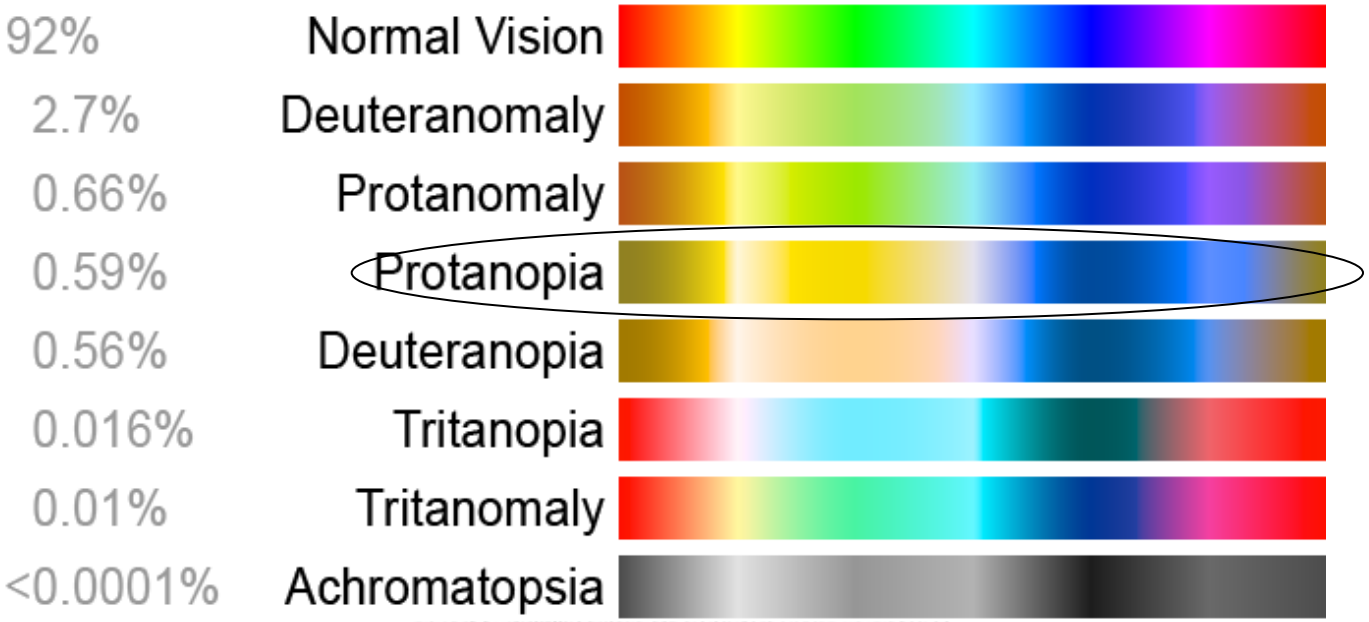
In males, a defective version of any one of these genes produces colorblindness.

Possible Inheritance of Colorblindness Allele



Copyright Pearson Prentice Hall

14–2 Human Chromosomes



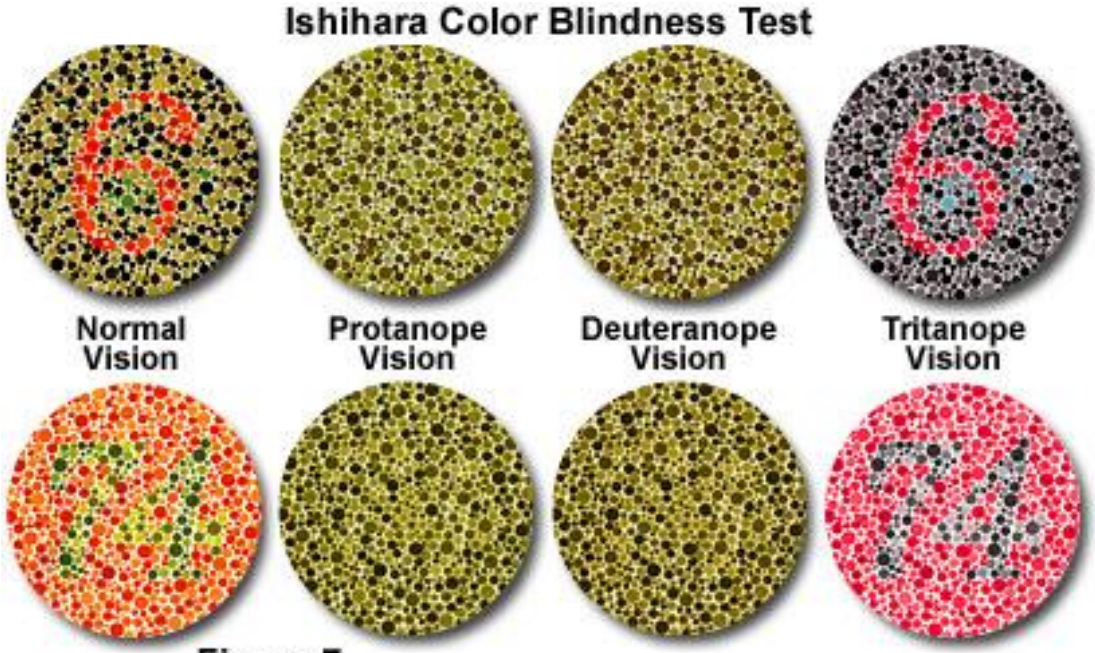


Figure 7

PEARSON Prentice Hall

Hemophilia

The X chromosome also carries genes that help control blood clotting. A recessive allele in either of these two genes may produce hemophilia.

In hemophilia, a protein necessary for normal blood clotting is missing.

Hemophiliacs can bleed to death from cuts and may suffer internal bleeding if bruised.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

Duchenne Muscular Dystrophy

Duchenne muscular dystrophy is a sex-linked disorder that results in the weakening and loss of skeletal muscle.

It is caused by a defective version of the gene that codes for a muscle protein.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

X-Chromosome Inactivation

British geneticist Mary Lyon discovered that in female cells, one X chromosome is randomly switched off.

This chromosome forms a dense region in the nucleus known as a Barr body.

Barr bodies are generally not found in males because their single X chromosome is still active.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

The most common error in meiosis occurs when homologous chromosomes fail to separate in Meiosis 1.

This is known as **nondisjunction**, which means, "not coming apart."

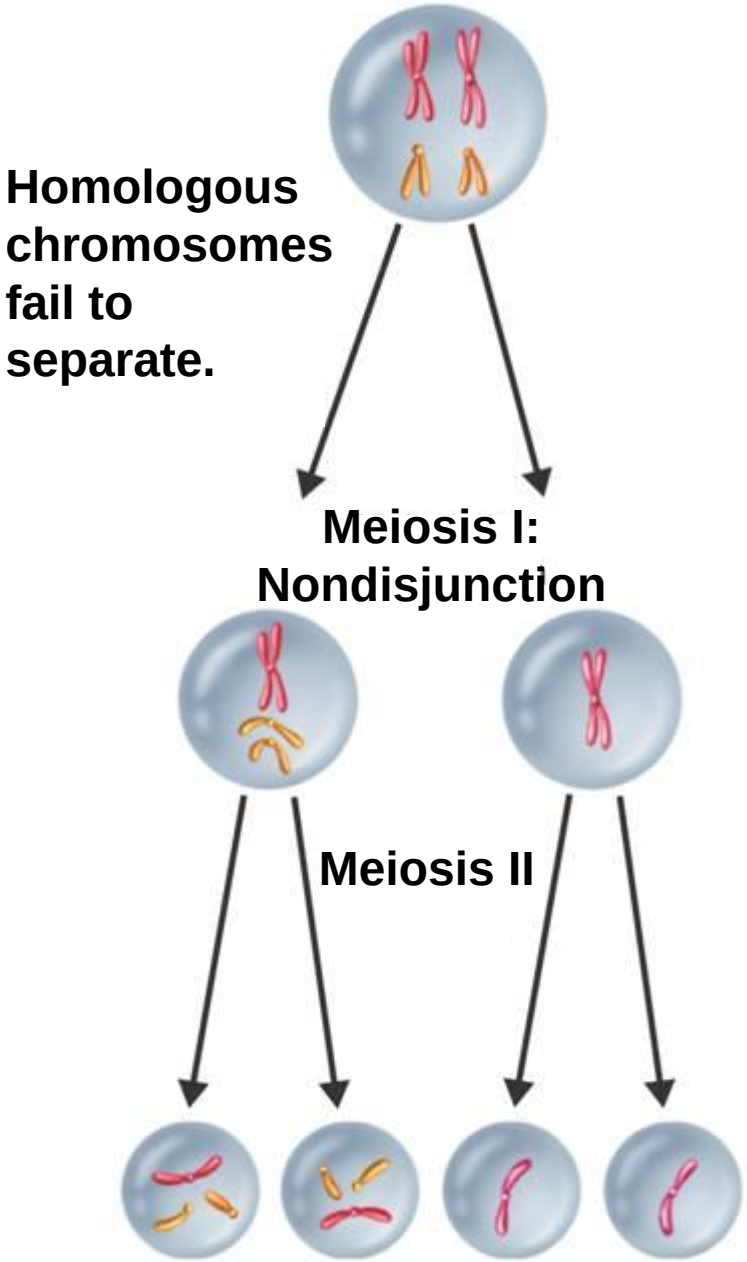
If nondisjunction occurs, abnormal numbers of chromosomes may find their way into gametes, and a disorder of chromosome numbers may result.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

movie
click to start

Nondisjunction



PEARSON Prentice Hall

Copyright Pearson Prentice Hall

Down Syndrome

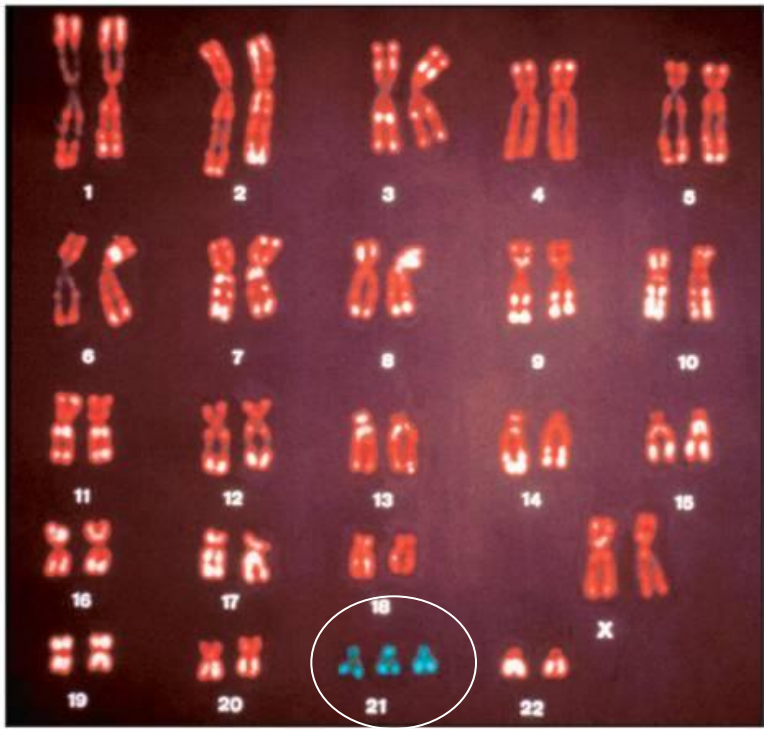
If two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with three copies of a chromosome.

Down syndrome involves three copies of chrome # 21.

Down syndrome produces mild to severe mental retardation. It is characterized by:

* increased susceptibility to many diseases

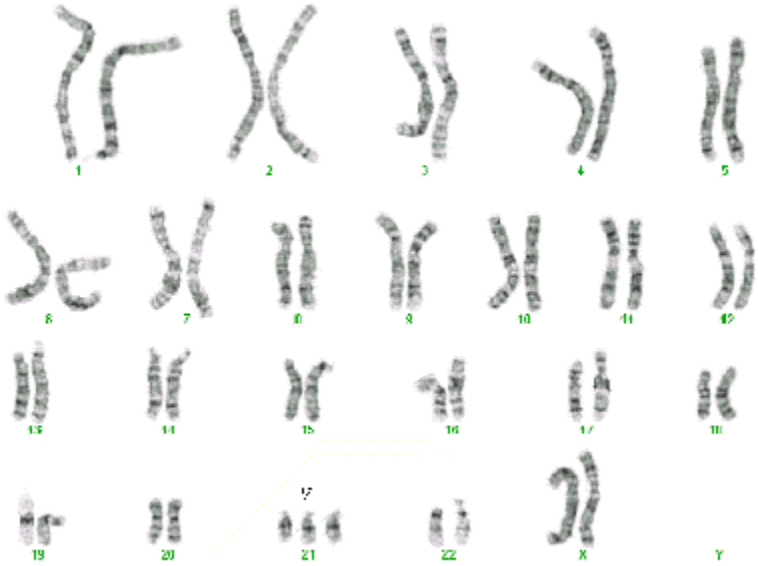
* higher frequency of some birth defects



PEARSON Prentice Hall

Copyright Pearson Prentice Hall

14–2 Human Chromosomes



PEARSON
Prentice Hall

Copyright Pearson

Sex Chromosome Disorders

In females, nondisjunction can lead to Turner's syndrome.

A female with Turner's syndrome usually inherits only one X chromosome (karyotype 45,X) or (45, XO)

Women with Turner's syndrome are sterile.

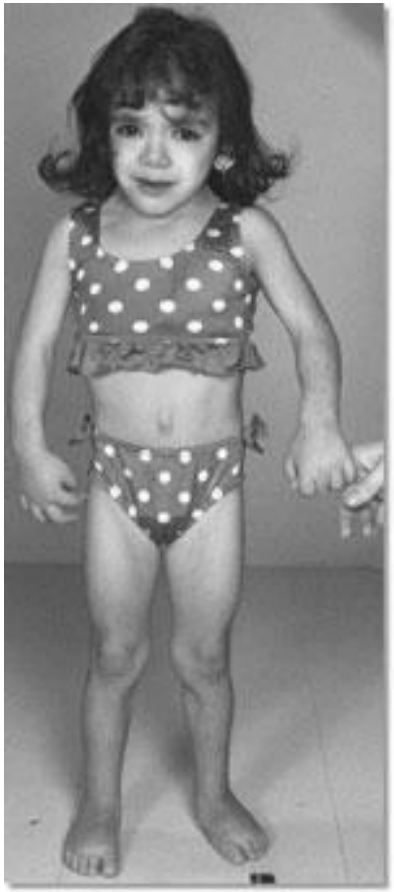
In males, nondisjunction causes Klinefelter's syndrome (karyotype 47,XXY).

The extra X chromosome interferes with meiosis and usually prevents these individuals from reproducing.

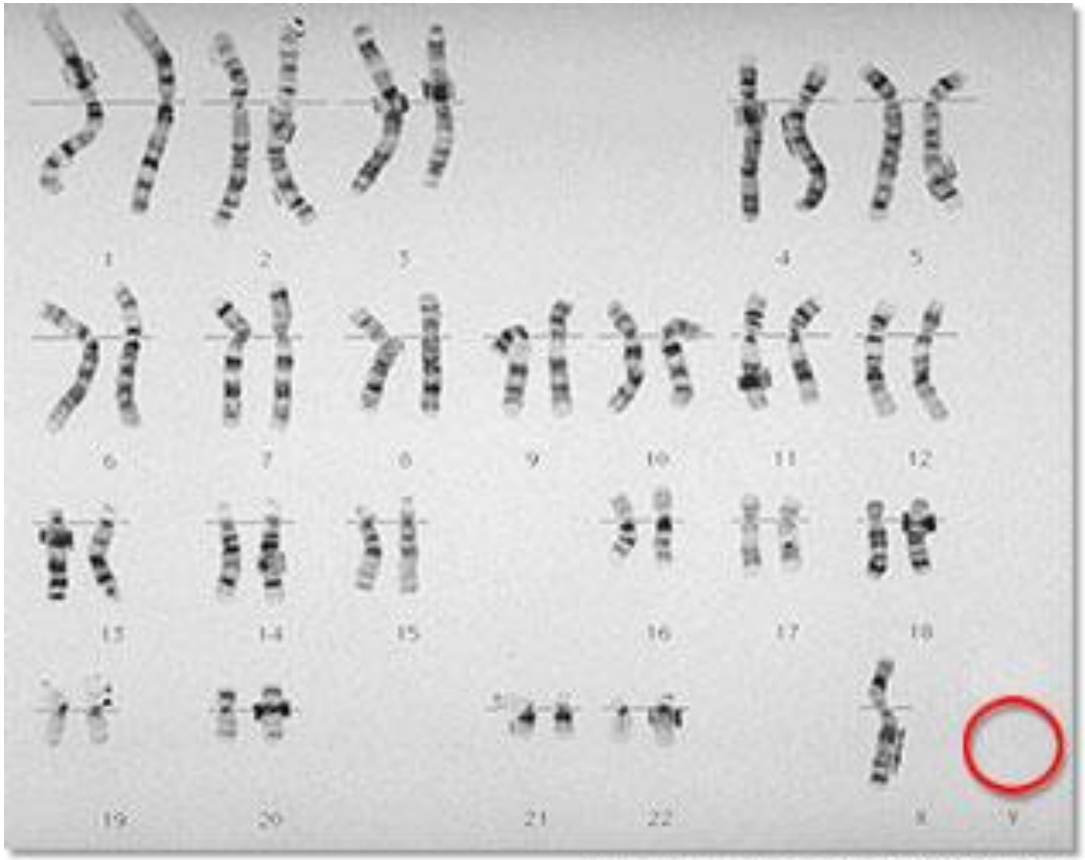
PEARSON Prentice Hall

Copyright Pearson Prentice Hall

Turner's Syndrome



medgen.genetics.utah.edu

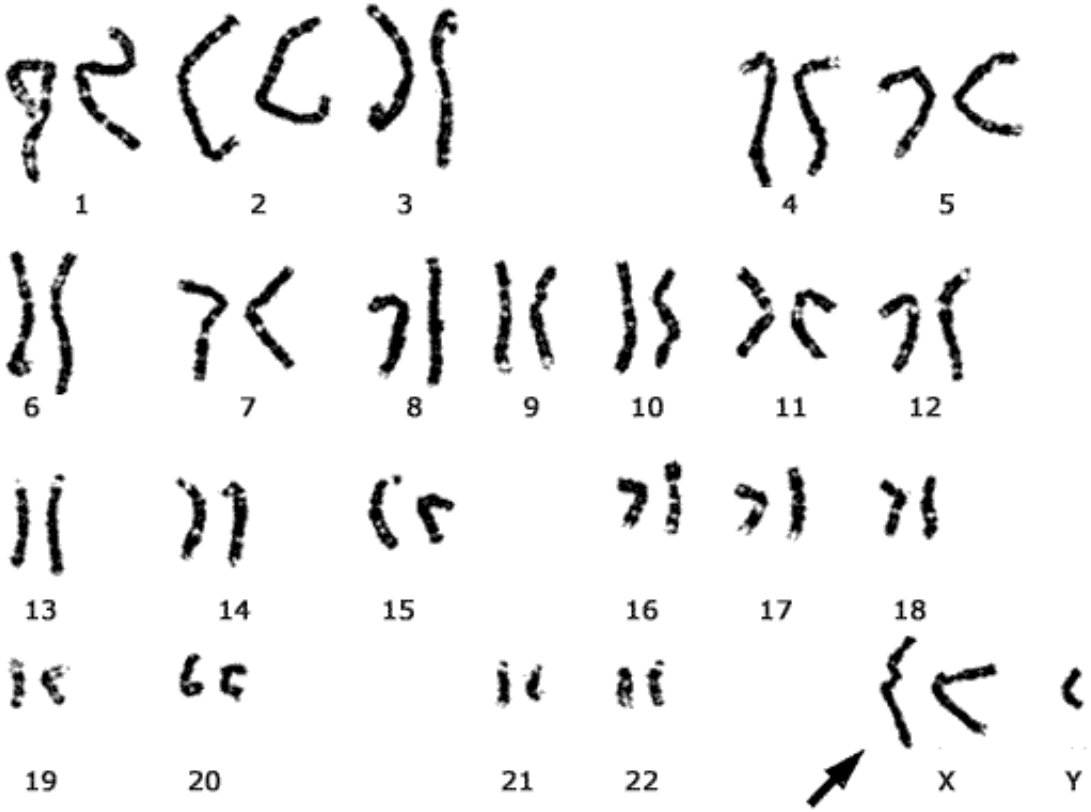


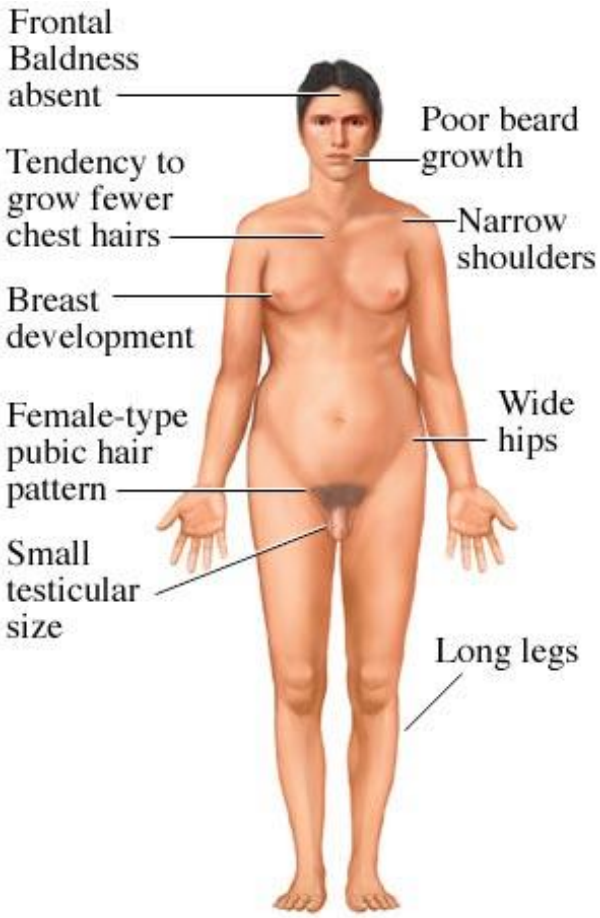
medgen.genetics.utah.edu

PEARSON Prentice Hall

Copyrigl

Klinefelter Syndrome





PEARSON Prentice Hall

Copyright Pearson Prentice Hall

14–2 Section QUIZ

Continue to:

Section QUIZ

- or -

Click to Launch:

ExamView® QuickTake

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

14–2 Section QUIZ

1 The average human gene consists of how many base pairs of DNA?

A a. 3000

b. 300

c. 20

d. 30,000

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

2 Which of the following genotypes indicates an individual who is a carrier for colorblindness?

a. X^CX

A b. X^CX^c

c. X^cY

d. X^CY

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

3 Colorblindness is much more common in males than in females because

A a. the recessive gene on the male's single X chromosome is expressed.

b. genes on the Y chromosome make genes on the X chromosome more active.

c. females cannot be colorblind.

d. colorblindness is dominant in males and recessive in females.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

4 The presence of a dense region in the nucleus of a cell can be used to determine the

A a. sex of an individual.

b. blood type of an individual.

c. chromosome number of an individual.

d. genotype of an individual.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall

5 Nondisjunction occurs during

A a. meiosis I.

b. mitosis.

c. meiosis II.

d. between meiosis I and II.

PEARSON Prentice Hall

Copyright Pearson Prentice Hall