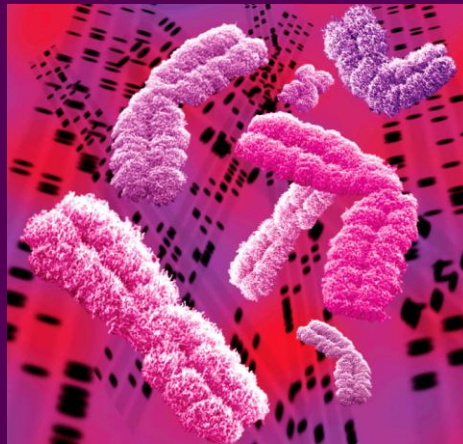


# BIOLOGY I

## Chapter 15: THE CHROMOSOMAL BASIS OF INHERITANCE



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Instructor

## Mendelian inheritance has its physical basis in the behavior of chromosomes

- **Chromosome theory of inheritance:** The biology principle stating that genes are located on chromosomes and that the behavior of chromosomes during meiosis accounts for inheritance patterns.
- Parallels observed between the behavior of chromosomes and the behavior of Mendel's proposed hereditary factors during sexual life cycles gave rise to the chromosome theory of inheritance.
  - Chromosomes and genes are both present in pairs in diploid cells.
  - Homologous chromosomes separate and alleles segregate during the process of meiosis.
  - Fertilization (union of gametes) restores the paired condition for both chromosomes and genes.

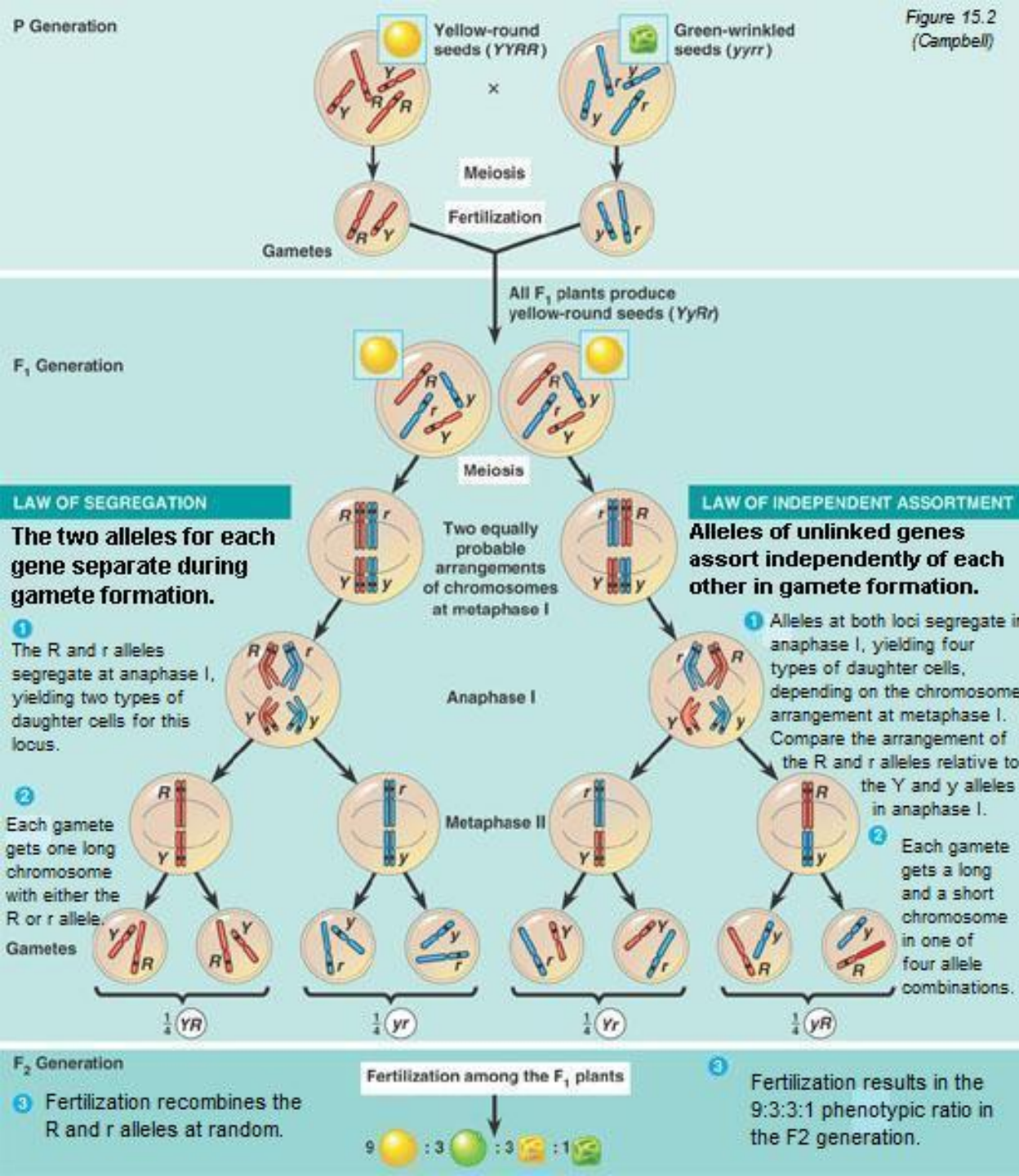
Figure 15.2 (Campbell)

Basis of Inheritance

**The chromosomal basis of Mendel's laws.** Correlation of the results of one of Mendel's dihybrid crosses with the behavior of chromosomes during meiosis.

**Law of segregation:** The two alleles for each gene separate during gamete formation. As an example, follow the fate of the long chromosomes (carrying *R* and *r*).

**Law of independent assortment:** Each pair of alleles assort independently of each other pair during gamete formation (it applies when genes for two characters are located on different pairs of homologous chromosomes). As an example, follow both the long and short chromosomes along both paths.





## Sex Determination in Humans and Sex-linked (X-linked) Genes

- An organism's sex is an inherited phenotypic character usually determined by the presence or absence of certain chromosomes.

- Humans and other mammals have an **X-Y system** in which sex normally is determined by the **presence or absence of a Y chromosome**.

- **Female: XX** (the person inherits two X chromosomes)
- **Male: XY** (the person inherits one X chromosome and one Y chromosome)

- Different systems of sex determination are found in birds, fishes, and insects.

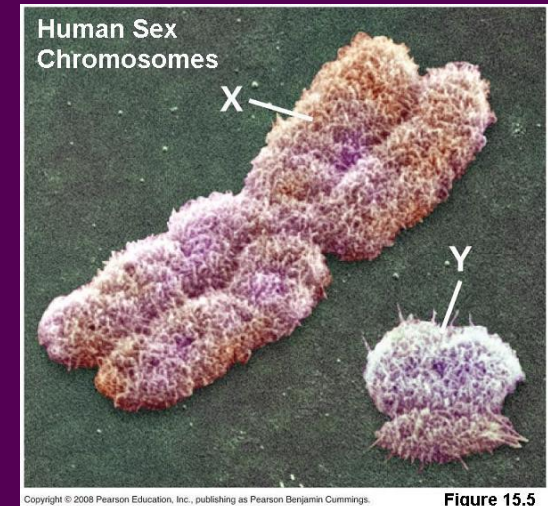


Figure 15.5

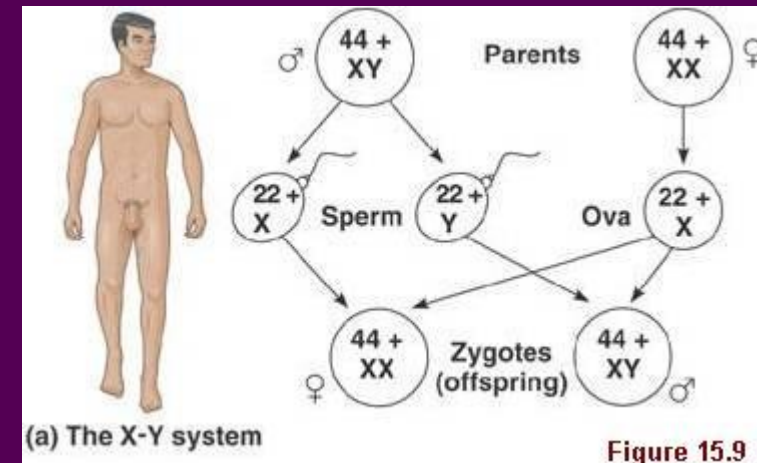
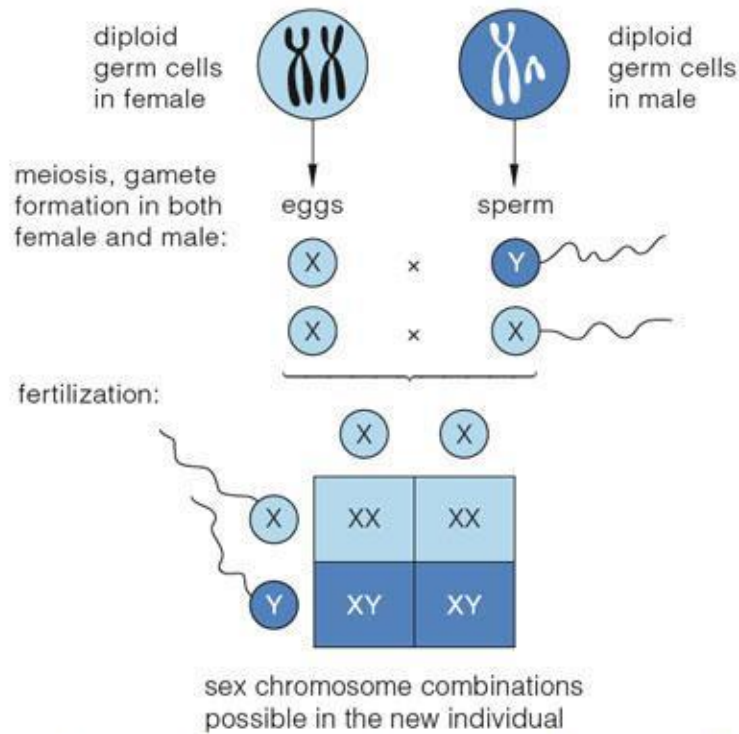


Figure 15.9

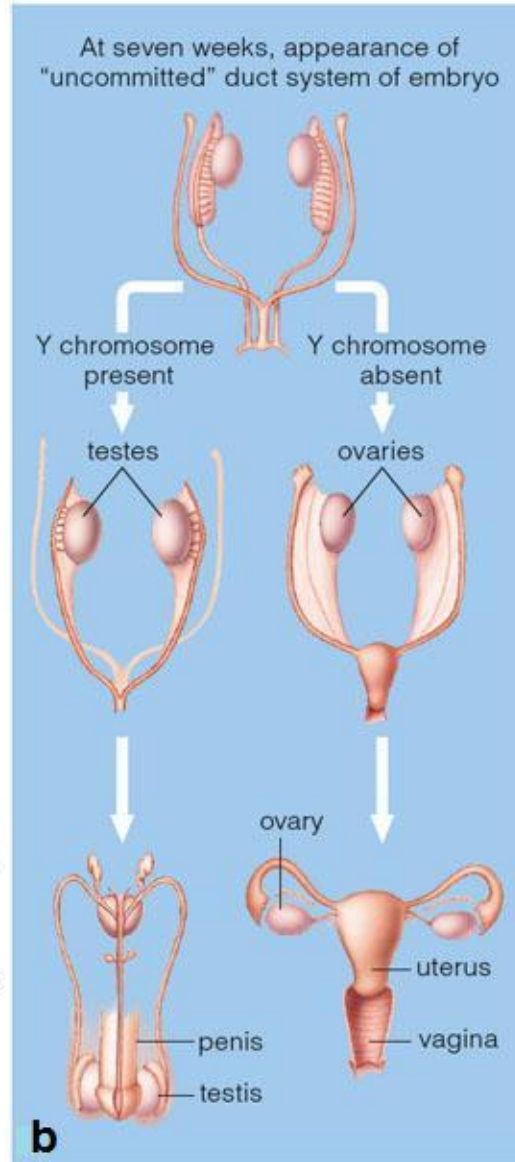
In mammals, the sex of an offspring depends on whether the sperm cell contains an X chromosome or a Y.

# Sex Determination in Humans

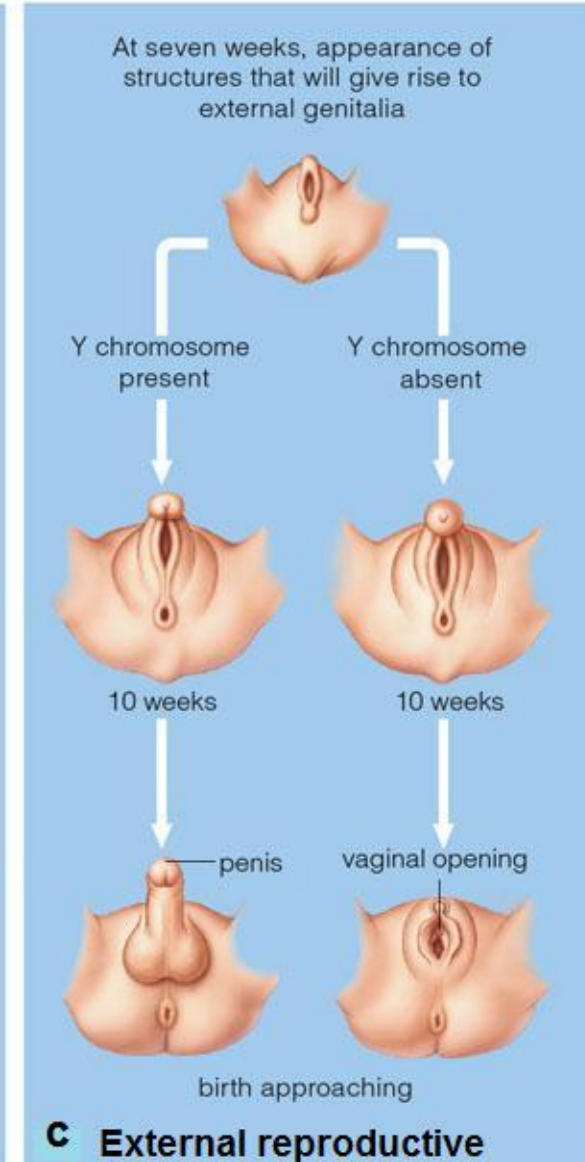


**a. Punnett square diagram showing the sex determination pattern in humans.**

**b. Early on, a human embryo is neither male nor female. Structures that can develop into male or female reproductive organs start forming.**



**b**



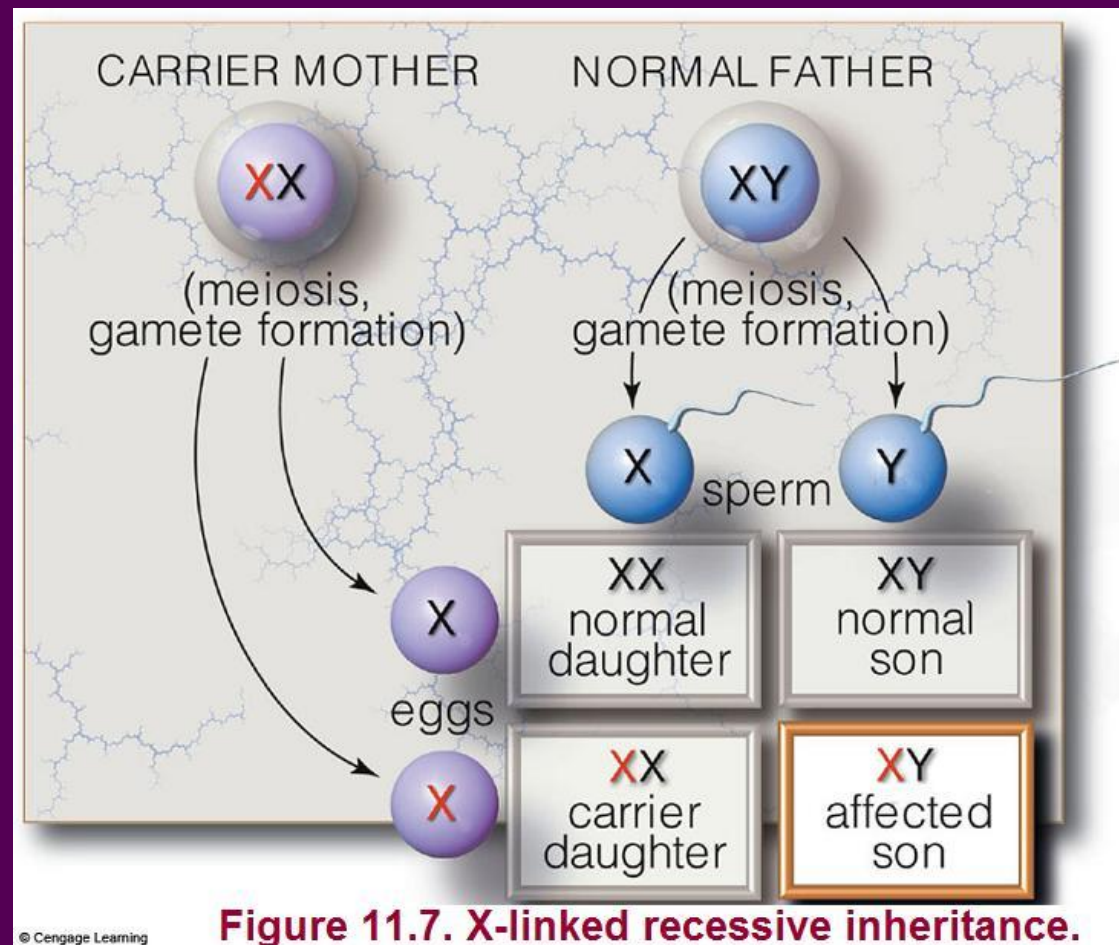
**c External reproductive organs in human embryos**

## Inheritance of Sex-Linked (X-Linked) Genes

- The **sex chromosomes** carry certain genes for traits that are unrelated to maleness or femaleness.
- A **sex-linked gene** is a gene located on either sex chromosome. An **X-linked gene** is a gene located only on the X chromosome.
- **Examples:** Recessive alleles causing color blindness, hemophilia, and Duchenne muscular dystrophy are carried on the X chromosome.
  - **Fathers** transmit such sex-linked (X-linked) alleles to **all daughters** but to no sons.
  - **Mothers** can pass sex-linked alleles to both **sons and daughters**.
  - Any **male** who inherits a sex-linked recessive allele **from his mother** will **express the trait**. For this reason, far more males than females have sex-linked recessive disorders.



## X-Linked Inheritance



- In this case, the mother carries a recessive allele on one of her X chromosomes (red).

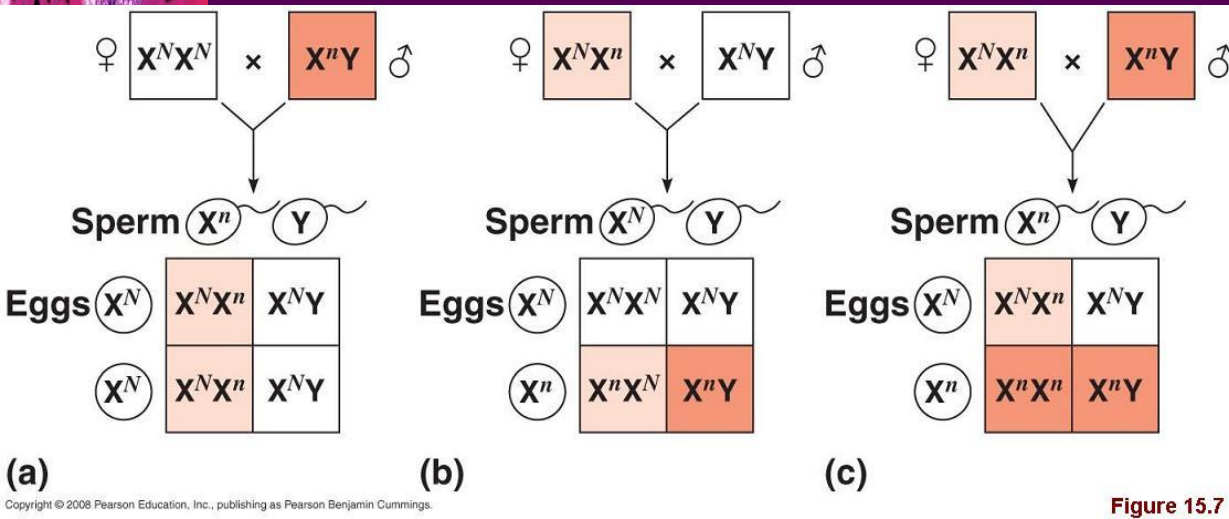


Figure 15.7

Superscript **N** = dominant allele for normal color vision on X chromosome.

Superscript **n** = recessive allele, with a mutation causing color blindness.

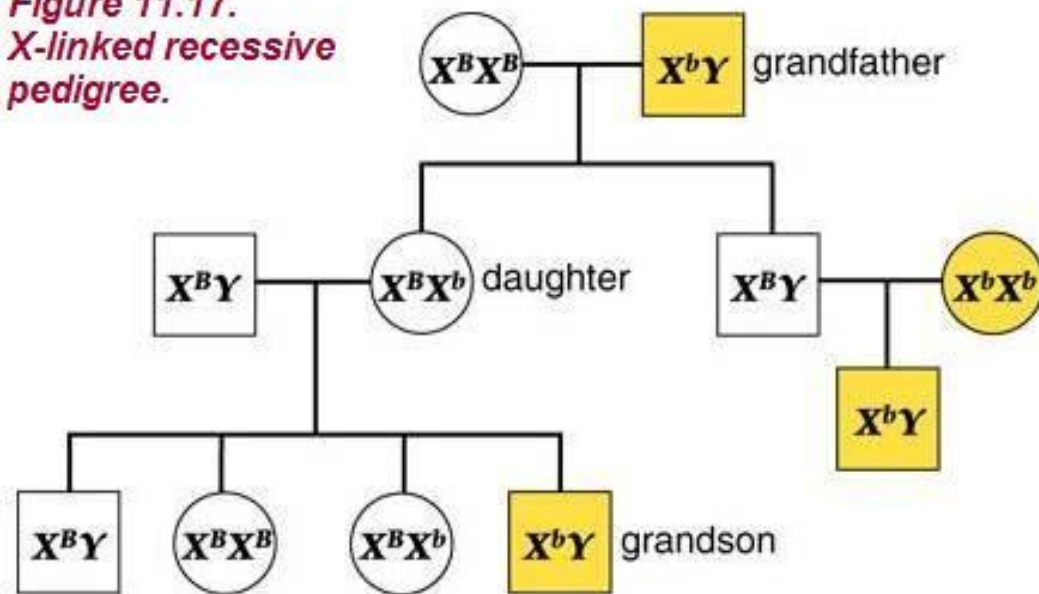
**White boxes** = unaffected individuals; **light orange boxes** = carriers; **dark orange boxes** = color-blind individuals.

### The transmission of sex-linked recessive traits.

- a) A *color-blind father* will transmit the mutant allele to **all daughters** but to **no sons**. When the *mother is a dominant homozygote*, the **daughters** will have the normal phenotype but will be **carriers** of the mutation.
- b) If a *carrier* mates with a *male who has normal color vision*, there is a **50%** chance that each **daughter will be a carrier** like her mother and a **50%** chance that each **son will have the disorder**.
- c) If a *carrier* mates with a *color-blind male*, there is a **50%** chance that each child born to them **will have the disorder**, regardless of sex. **Daughters** who have normal color vision will be **carriers**, whereas **sons** who have normal color vision **will be free** of the recessive allele.



**Figure 11.17.**  
**X-linked recessive pedigree.**



Key	
$X^B X^B$	= Unaffected female
$X^B X^b$	= Carrier female
$X^b X^b$	= Color-blind female
$X^B Y$	= Unaffected male
$X^b Y$	= Color-blind male

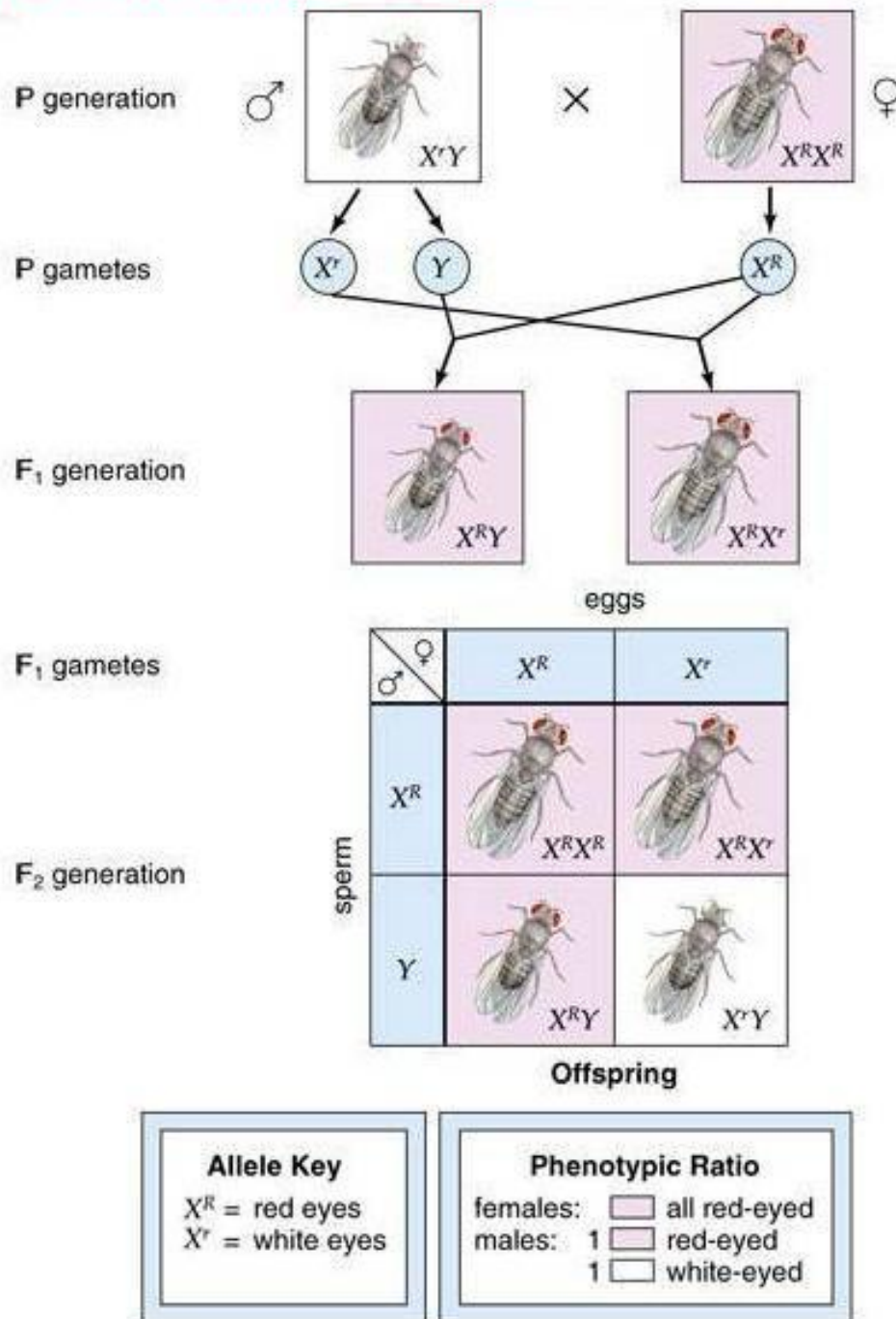
### X-linked Recessive Disorders

- More males than females are affected.
- An affected son can have parents who have the normal phenotype.
- For a female to have the characteristic, her father must also have it. Her mother must have it or be a carrier.
- The characteristic often skips a generation from the grandfather to the grandson.
- If a woman has the characteristic, all of her sons will have it.

## X-linked Recessive Pedigree

- This pedigree for **color blindness** exemplifies the inheritance pattern of an **X-linked recessive disorder**.
- The list gives various ways of recognizing the X-linked recessive pattern of inheritance.

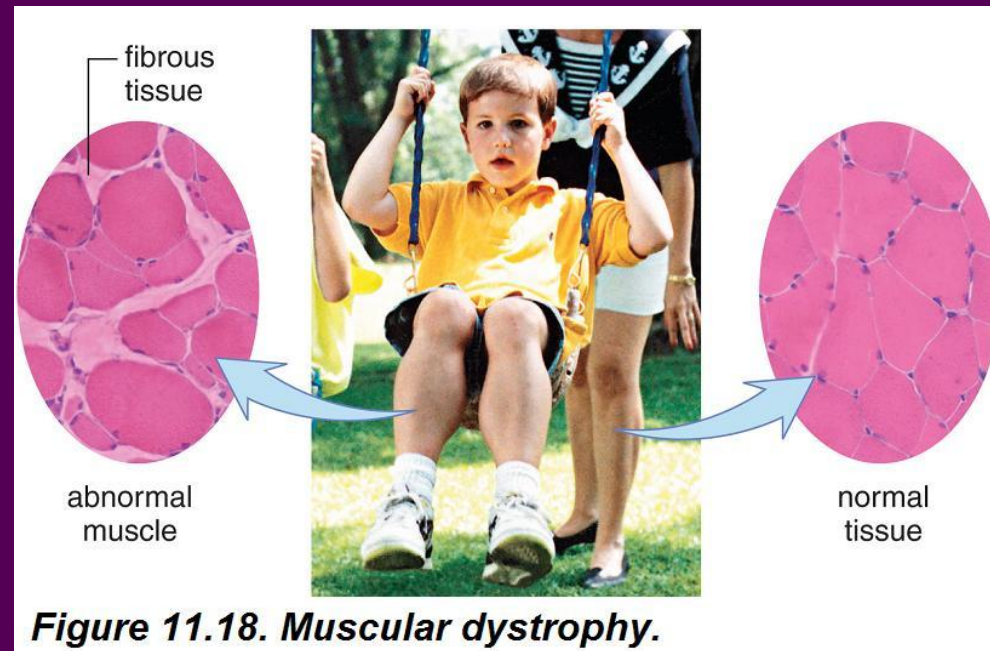
Figure 11.16. X-linked inheritance.



## Sex-linked Inheritance

- Once researchers deduced that the alleles for red/white eye color are on the X chromosome in *Drosophila* (fruit fly), they were able to explain their experimental results. Males with white eyes in the F<sub>2</sub> inherit the recessive allele only from the female parent; they receive a Y chromosome lacking the gene for eye color from the male parent.

## Human X-Linked Disorders



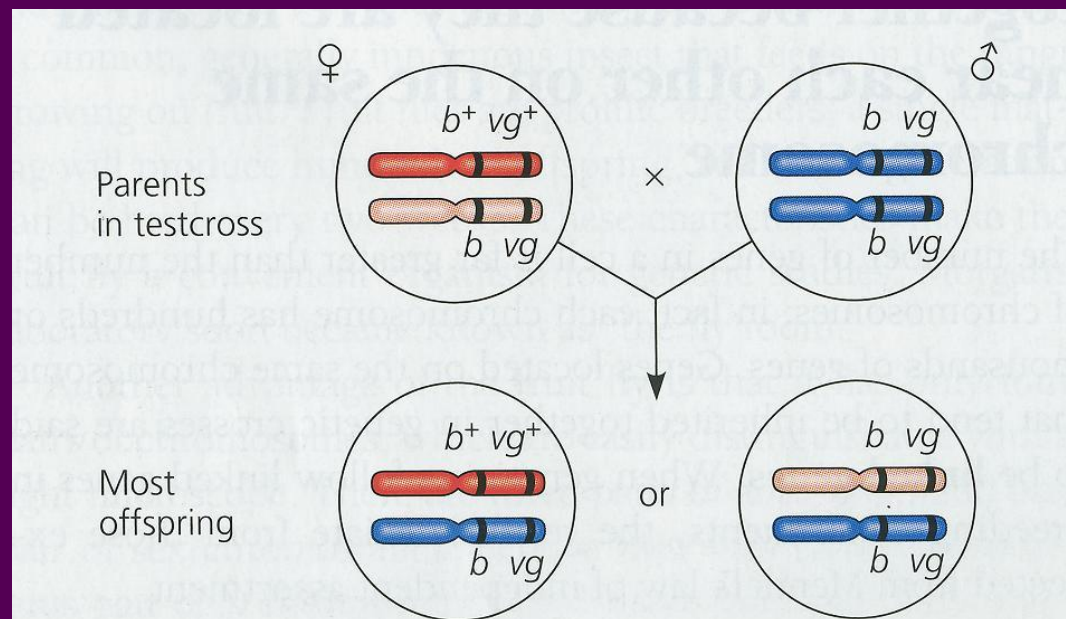
- In muscular dystrophy, an X-linked recessive disorder, fibrous tissue develops as muscles waste away, due to lack of the protein dystrophin. The most common form, **Duchenne muscular dystrophy**, occurs in about 1 out of every 3,600 male births. Symptoms include waddling gait, toe walking, frequent falls, and difficulty in rising. Muscle weakness intensifies until the individual is confined to a wheelchair. Death usually occurs by age 20; therefore, affected males are rarely fathers. The recessive allele remains in the population through passage from carrier mother to carrier daughter.

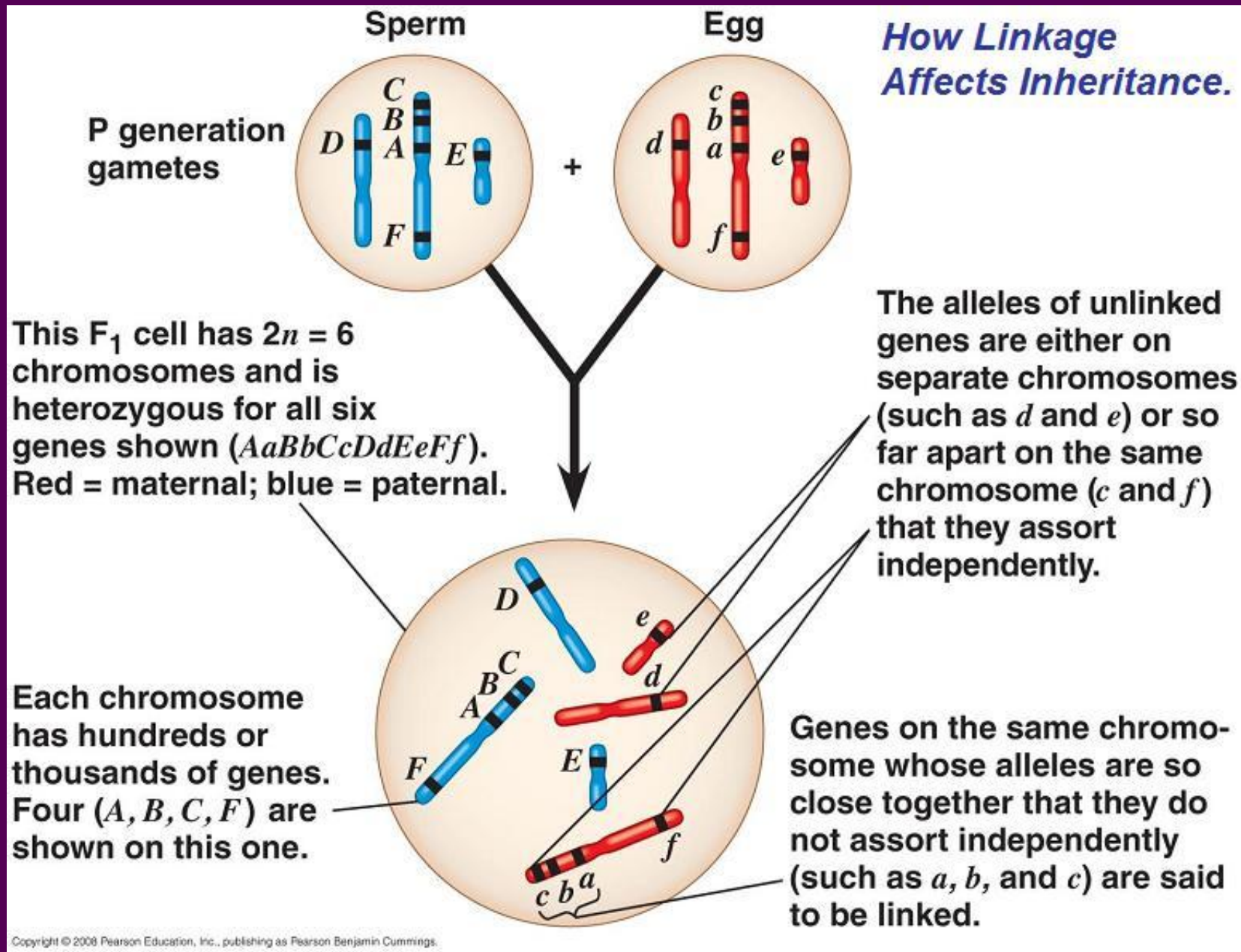


# Linked Genes Tend to be Inherited Together

- **Linked genes:** Genes located close enough together on the same chromosome to be usually **inherited together** (they do not assort independently).
- The alleles of **unlinked genes** are either on separate chromosomes or so far apart on the same chromosome that they assort independently.

\* Note the distinction between the terms *sex-linked gene*, referring to a single gene on a sex chromosome, and *linked genes*, referring to two or more genes on the same chromosome that tend to be inherited together.







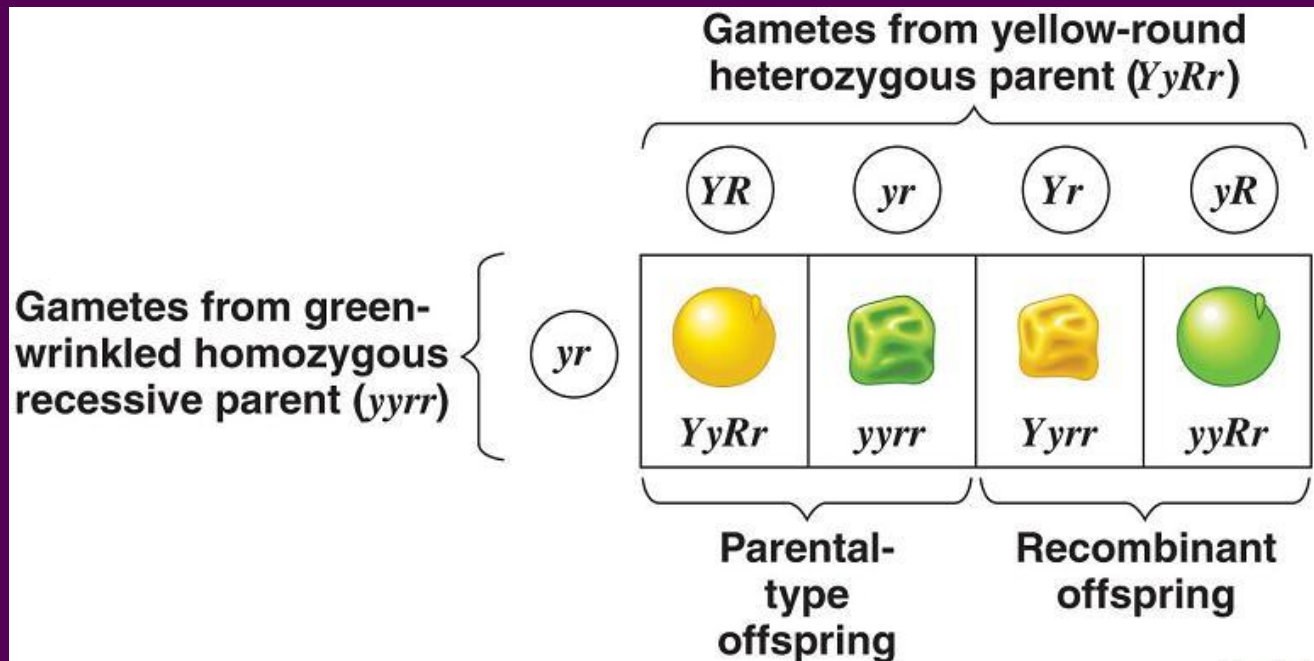
## Genetic Recombination and Linkage

- Any process that leads to new gene combinations is called **genetic recombination**.
- **Crossing-over**, a process in which genetic material (DNA) is exchanged between paired, homologous chromosomes, can cause recombinant gametes and recombinant phenotypes to occur.
- Recombinant offspring exhibit new combinations of traits inherited from two parents.
- **Unlinked genes** exhibit a 50% frequency of recombination because of the independent assortment of chromosomes and random fertilization.
- **Linked genes** exhibit recombination frequencies **less than 50%**, even with crossing over between nonsister chromatids during meiosis I.



## Recombination of Unlinked Genes: *Independent Assortment of Chromosomes*

- Notice in this Punnett square that one-half of the offspring are expected to inherit a phenotype that matches one of the parental phenotypes. These offspring are called **parental types**. But two nonparental phenotypes are also found among the offspring. Because these offspring have new combinations of seed shape and color, they are called **recombinant types**, or **recombinants**, for short.



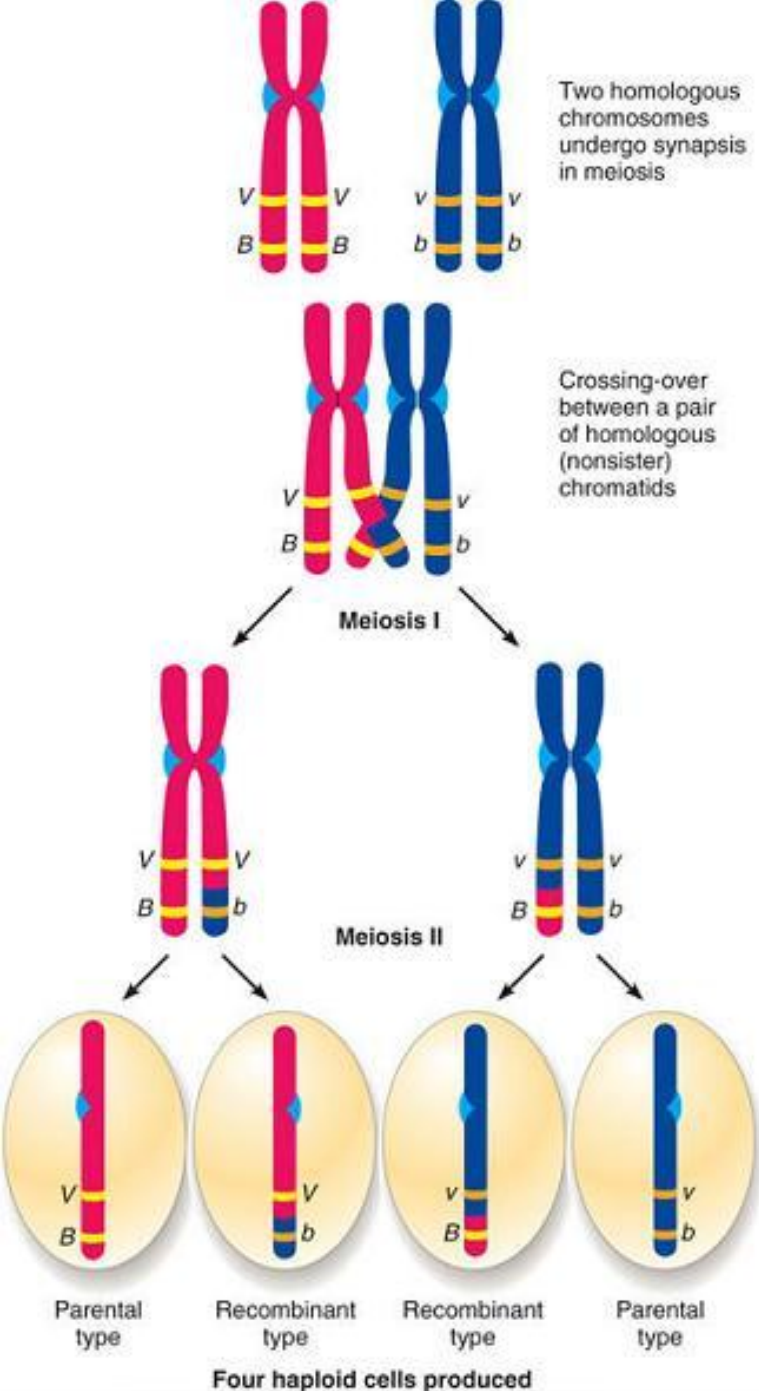


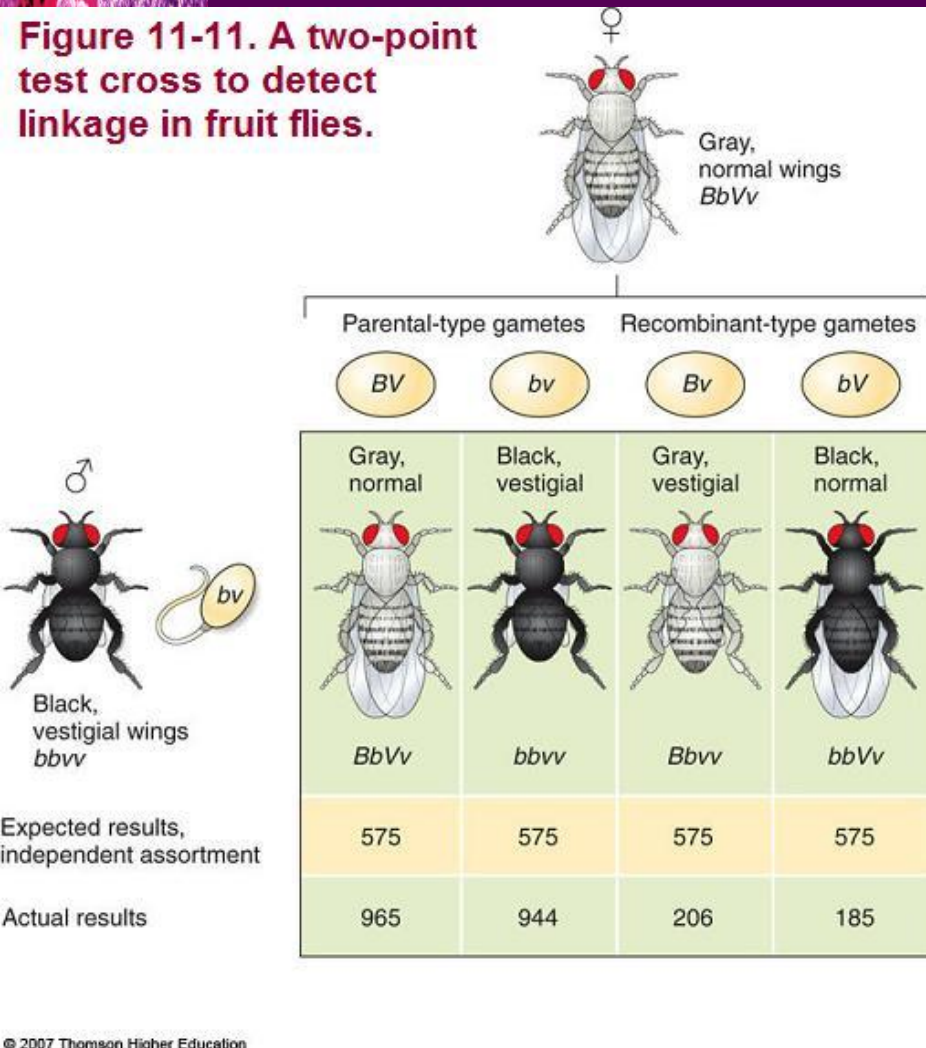
Figure 11-12. Crossing-over

## Recombination of Linked Genes: Crossing Over

- When homologous chromosomes are in synapsis (during prophase of meiosis I), the nonsister chromatids **exchange genetic material**. This **crossing-over** is the mechanism that accounts for the recombination of **linked genes**.
- Following **crossing-over**, recombinant chromosomes result. Recombinant chromosomes contribute to recombinant gametes.
- Genes located **far apart** on a chromosome have a **greater probability of being separated by crossing-over** than do genes that are closer together.

# A two-point test cross to detect linkage in fruit flies

**Figure 11-11. A two-point test cross to detect linkage in fruit flies.**



- **Linkage** can be recognized when an excess of parental-type offspring and a deficiency of recombinant-type offspring are produced in a two-point test cross (alleles of two loci involved).
- Fruit flies with gray, normal wings ( $BbVv$ ) are crossed with flies that have black, vestigial wings ( $bbvv$ ). If the alleles for color and wing shape are not linked (i.e., the alleles assort independently), the offspring will consist of an equal number of each of four phenotypes (yellow row).
- In the 2300 offspring (bottom row) of an actual cross, about 1909 of the offspring belong to each of the two parental crosses (83% total), and 391 belong to each of the two recombinant classes (17% total). Thus, loci for wing length and body color are **linked** on a homologous chromosome pair.

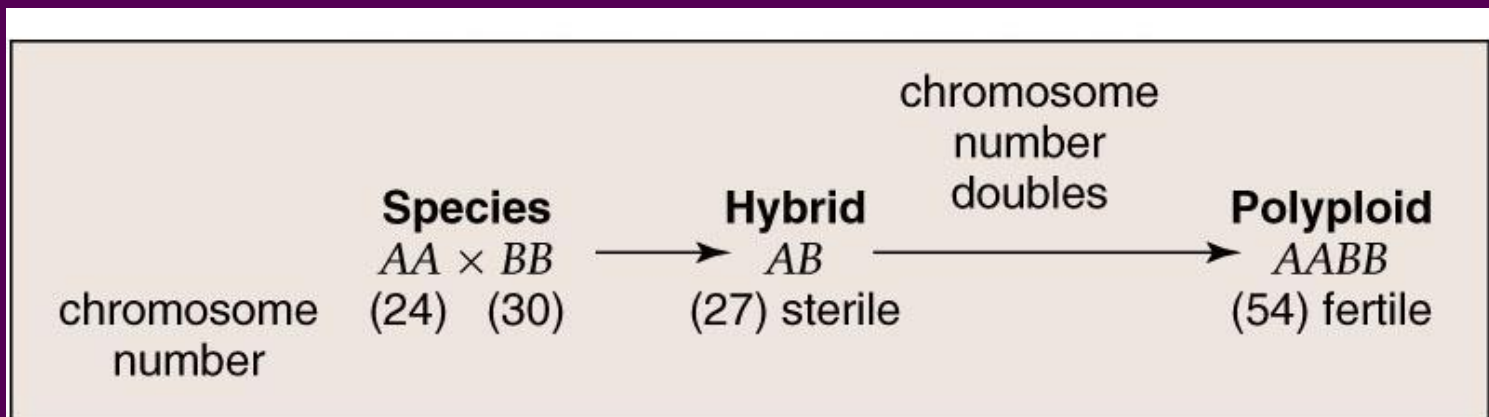


## Alterations of Chromosome Number or Structure

- Physical and chemical **disturbances and errors during meiosis** can damage chromosomes, or alter their number in a cell.
- Alterations of chromosome number or structure cause some **genetic disorders**.
- Large-scale chromosomal alterations often lead to spontaneous abortion (**miscarriage**) of a fetus, and individuals born with these types of genetic defects commonly exhibit various **developmental disorders**.
- Plants tend to tolerate such genetic defects to a greater extent than animals.

## Changes in Chromosome Number

- **Polyploidy:** A chromosomal alteration in which the organism possesses more than two complete sets of chromosomes, for example, *triploidy* ( $3n$ ) or *tetraploidy* ( $4n$ ). It is the result of an accident in cell division.
  - It is not often seen in animals.
  - In plants, it is a major evolutionary mechanism.
    - Wheat, corn, cotton, sugarcane, watermelons, strawberries, bananas, apples.
    - The flowers and fruits of many polyploid plants are larger than their diploid counterparts.
  - Polyploidy generally arises following hybridization.

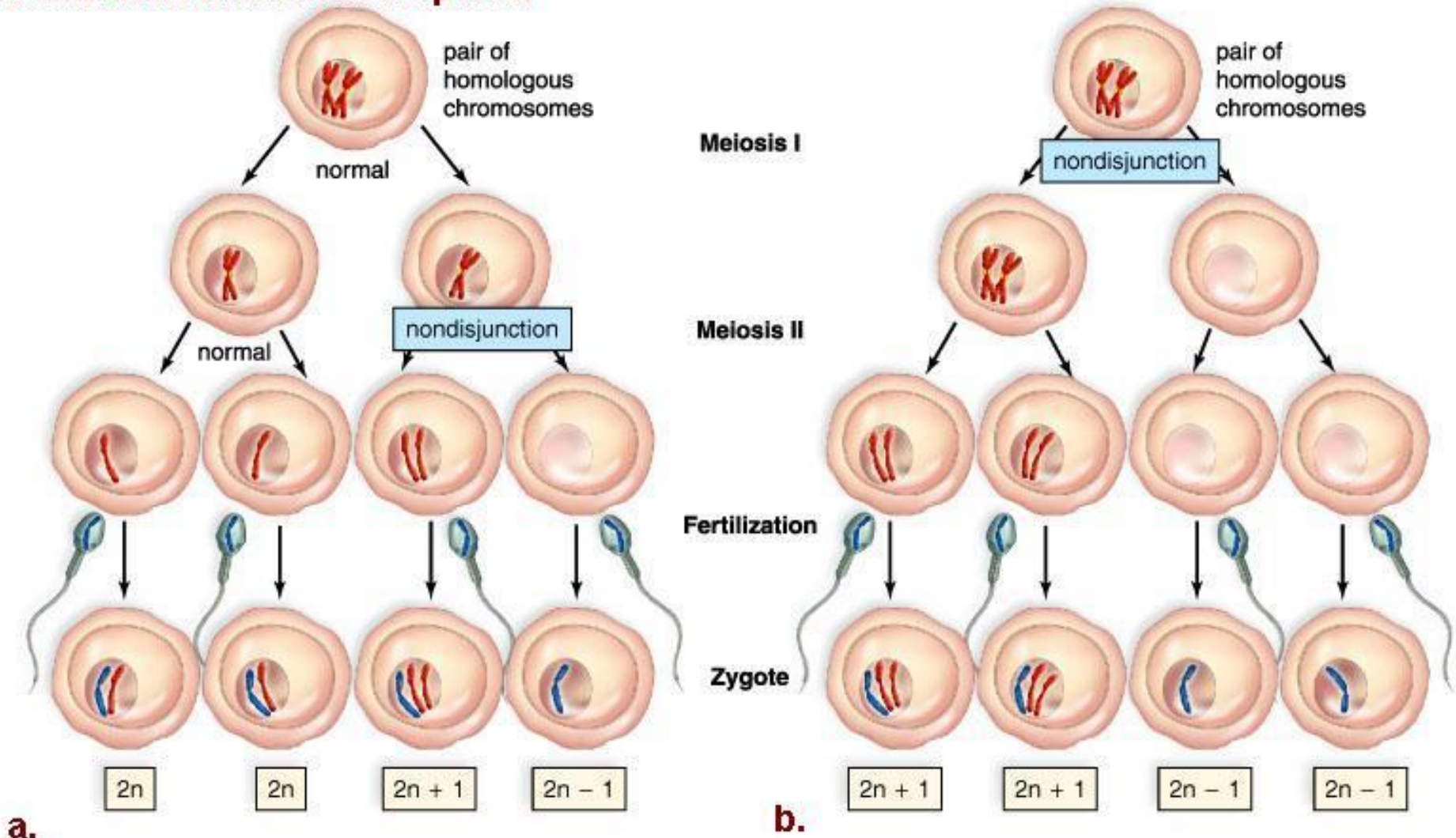


# Changes in Chromosome Number

- **Aneuploidy:** When an organism has *more or less* than the normal number of chromosomes.
  - **Disomy** is the normal state: two of each kind of chromosome.
  - **Monosomy ( $2n - 1$  chromosomes):** Occurs when an individual has only one of a particular type of chromosome.
  - **Trisomy ( $2n + 1$  chromosomes):** Occurs when an individual has three of a particular type of chromosome.
  - The cause is **nondisjunction**; it occurs during meiosis I when both members of a homologous pair of chromosomes go into the same daughter cell, or during meiosis II when the sister chromatids fail to separate and both daughter chromosomes go into the same gamete.



**Figure 12.10 - Nondisjunction of chromosomes during oogenesis, followed by fertilization with normal sperm.**



## Chromosome Abnormalities: *Aneuploidies*



**Figure 15.16.  
Down Syndrome.**

It is the result of an extra chromosome 21, so that each body cell has a total of 47 chromosomes.

The child exhibits the facial features characteristic of Down syndrome. The karyotype shows trisomy 21, the cause of this disorder.



- Persons with **Down syndrome**, or **trisomy 21**, have an extra chromosome 21.
- Common characteristics of the syndrome include a wide, rounded face, a fold on the upper eyelids, short stature, heart defects, susceptibility to respiratory infection, mental retardation, difficulty to speak.
- It affects approximately one out of every 700 children born in the United States.
- Some live to middle age or beyond.



# Chromosome Abnormalities: *Aneuploidies*

TABLE 16-2

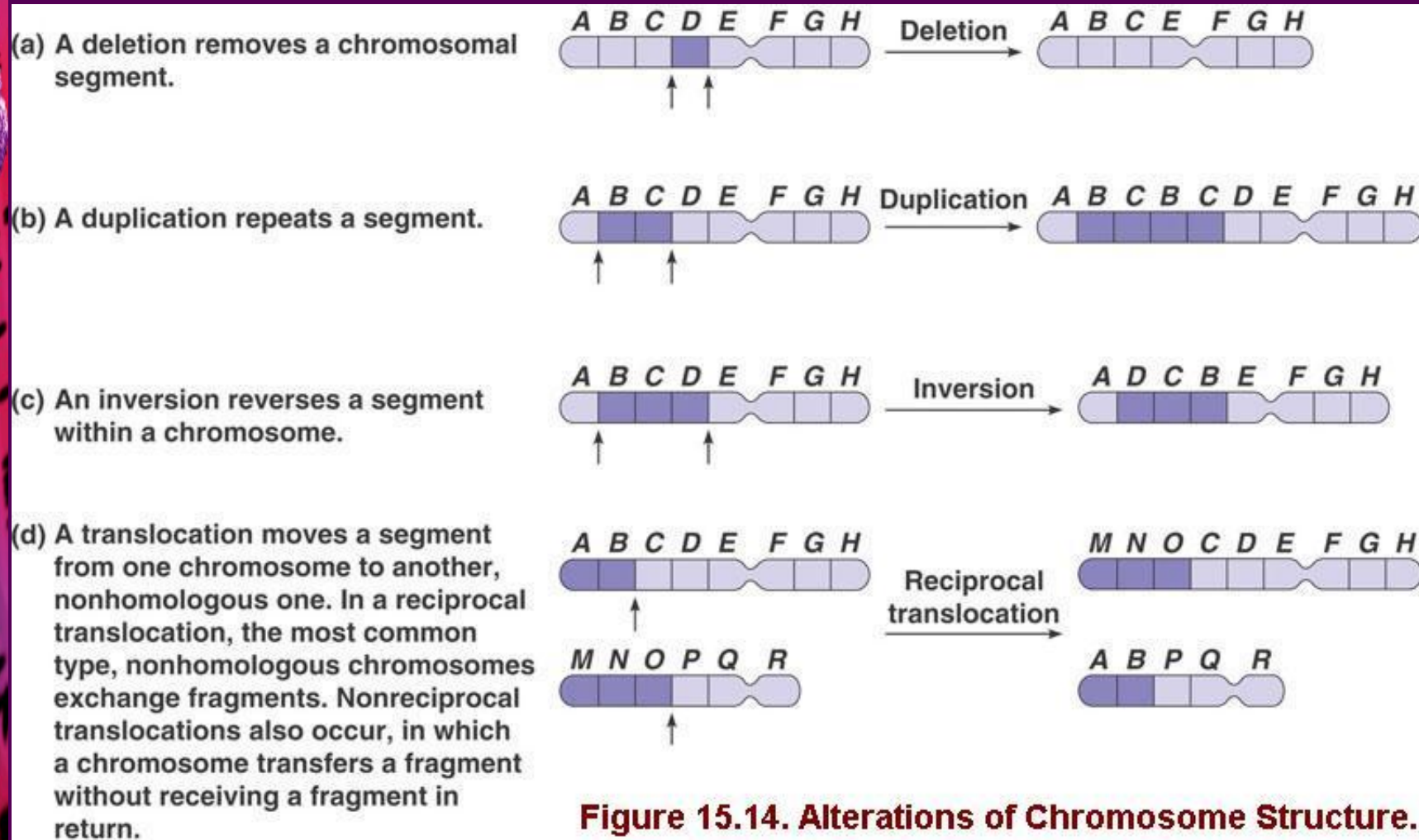
## Chromosome Abnormalities: Disorders Produced by Aneuploidies

Karyotype	Common Name	Clinical Description
Trisomy 13	Patau syndrome	Multiple defects, with death typically by age 3 months.
Trisomy 18	Edwards syndrome	Ear deformities, heart defects, spasticity, and other damage; death typically by age 1 year, but some survive much longer.
Trisomy 21	Down syndrome	Overall frequency is about 1 in 800 live births. Most conceptions involving true trisomy occur in older (age 35+) mothers, but translocation resulting in the equivalent of trisomy is not age-related. Trisomy 21 is characterized by a fold of skin above the eye, varying degrees of mental retardation, short stature, protruding furrowed tongue, transverse palmar crease, cardiac deformities, and increased risk of leukemia and Alzheimer's disease.
X0	Turner syndrome	Short stature, webbed neck, sometimes slight mental retardation; ovaries degenerate in late embryonic life, leading to rudimentary sexual characteristics; gender is female; no Barr bodies.
XXY	Klinefelter syndrome	Male with slowly degenerating testes, enlarged breasts; one Barr body per cell.
XYY	XYY karyotype	Many males have no unusual symptoms; others are unusually tall, with heavy acne, and some tendency to mild mental retardation.
XXX	Triplo-X	Despite three X chromosomes, usually fertile females with normal intelligence; two Barr bodies per cell.



# Alterations of Chromosome Structure

- **Errors** in meiosis or **damaging agents** such as radiation can cause breakage of a chromosome.
- Four types of changes in chromosome structure are:
  - **Deletion:** **Loss** of a chromosomal **fragment** through breakage; consequently, certain genes are missing in the chromosome.
  - **Duplication:** A gene segment is **repeated** several to many hundreds or thousands of times.
  - **Inversion:** Reattachment of a chromosomal fragment to the original chromosome but in the **reverse direction**.
  - **Translocation:** Attachment of a chromosomal fragment to a **nonhomologous chromosome**. In a **reciprocal translocation**, two nonhomologous chromosomes exchange segments.



**Figure 15.14. Alterations of Chromosome Structure.**

- Figure 15.15. Alterations of chromosome structure.** Vertical arrows indicate breakage points. Dark purple highlights the chromosomal parts affected by the rearrangements.

## Translocation associated with chronic myelogenous leukemia (CML)

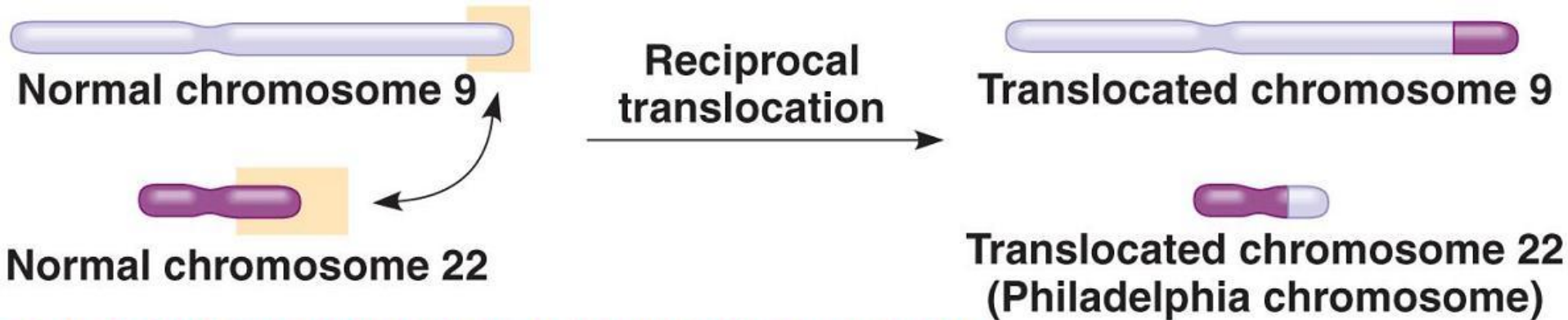


Fig. 15.17. Translocation associated with chronic myelogenous leukemia (CML). Copyright © 2008 Pearson Education, Inc., publishing as Pearson Benjamin Cummings.

- The cancerous cells in nearly all **CML** patients contain an **abnormally short chromosome 22**, the so-called *Philadelphia chromosome*, and an **abnormally long chromosome 9**. These altered chromosomes result from the translocation shown here, which presumably occurred in a single white blood cell precursor undergoing mitosis and was then passed along to all descendant cells.



Table 11.1 Examples of Human Genetic Disorders and Genetic Abnormalities

Disorder or Abnormality	Main Symptoms	Disorder or Abnormality	Main Symptoms
<b>Autosomal recessive inheritance</b>		<b>X-linked recessive inheritance</b>	
Albinism	Absence of pigmentation	Androgen insensitivity syndrome	XY individual but having some female traits; sterility
Hereditary methemoglobinemia	Blue skin coloration	Red–green color blindness	Inability to distinguish among some or all shades of red and green
Cystic fibrosis	Abnormal glandular secretions leading to tissue, organ damage	Fragile X syndrome	Mental impairment
Ellis–van Creveld syndrome	Dwarfism, heart defects, polydactyly	Hemophilia	Impaired blood clotting ability
Fanconi anemia	Physical abnormalities, bone marrow failure	Muscular dystrophies	Progressive loss of muscle function
Galactosemia	Brain, liver, eye damage	X-linked anhidrotic dysplasia	Mosaic skin (patches with or without sweat glands); other effects
Phenylketonuria (PKU)	Mental impairment		
Sickle-cell anemia	Adverse pleiotropic effects on organs throughout body		
<b>Autosomal dominant inheritance</b>		<b>Changes in chromosome structure</b>	
Achondroplasia	One form of dwarfism	Chronic myelogenous leukemia (CML)	Overproduction of white blood cells in bone marrow; organ malfunctions
Camptodactyly	Rigid, bent fingers	Cri-du-chat syndrome	Mental impairment; abnormally shaped larynx
Familial hypercholesterolemia	High cholesterol levels in blood; eventually clogged arteries		
Huntington's disease	Nervous system degenerates progressively, irreversibly		
Marfan syndrome	Abnormal or no connective tissue		
Polydactyly	Extra fingers, toes, or both		
Progeria	Drastic premature aging		
Neurofibromatosis	Tumors of nervous system, skin		
		<b>Changes in chromosome number</b>	
		Down syndrome	Mental impairment; heart defects
		Turner syndrome (XO)	Sterility; abnormal ovaries, abnormal sexual traits
		Klinefelter syndrome	Sterility; mild mental impairment
		XXX syndrome	Minimal abnormalities
		XYY condition	Mild mental impairment or no effect



## Testing a fetus for genetic disorders

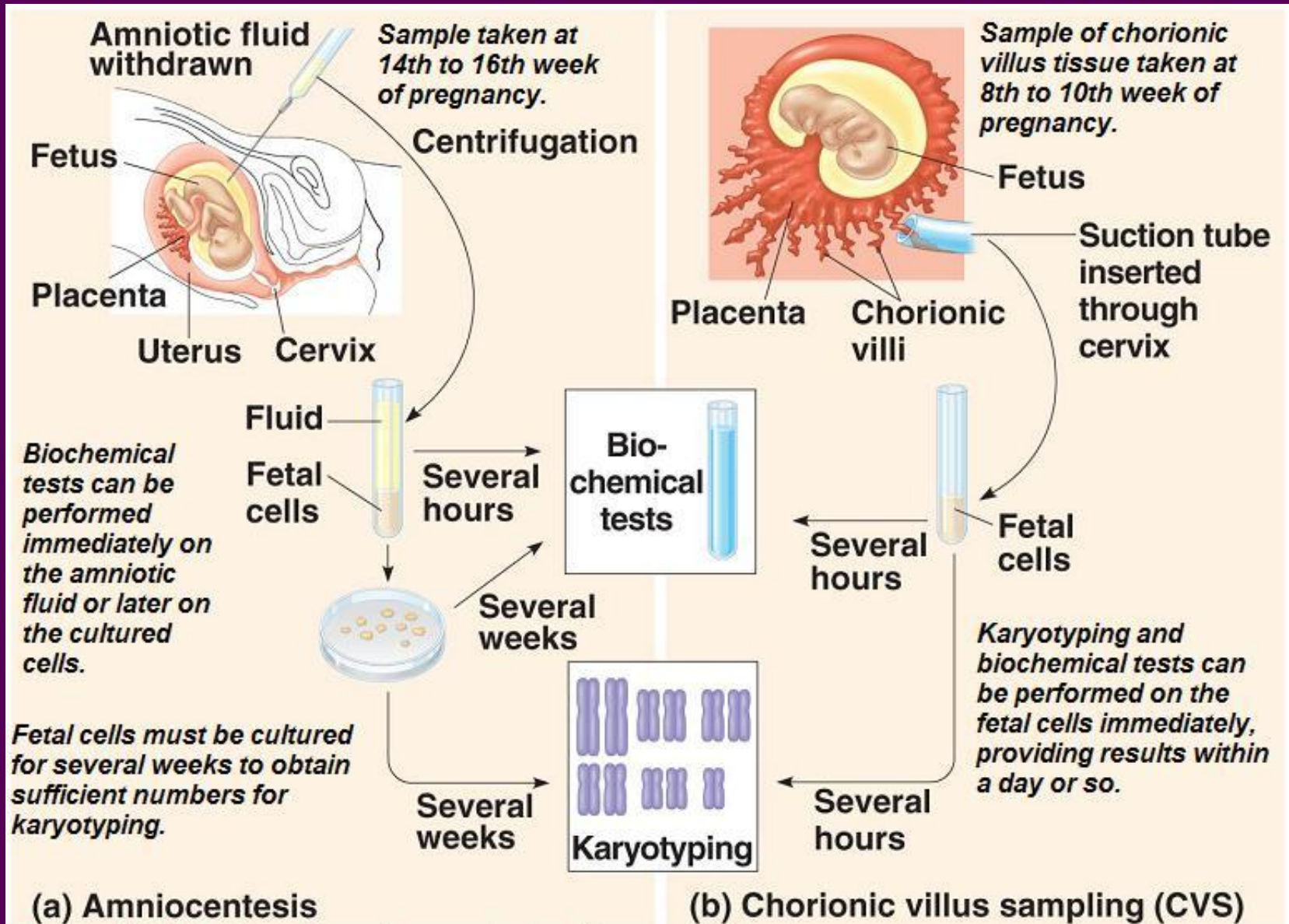
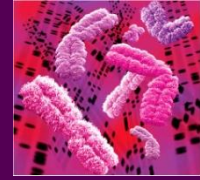


Figure 14.18. Testing a fetus for genetic disorders.



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