Ch. 12 Warm-Up

1. A white-eyed female fruit-fly is mated with a red-eyed male. What genotypes and phenotypes do you predict for the offspring?

2. Neither Tim nor Rhoda has Duchenne muscular dystrophy (X-linked recessive disorder), but their firstborn son has it. What is the probability their 2nd child will have it?

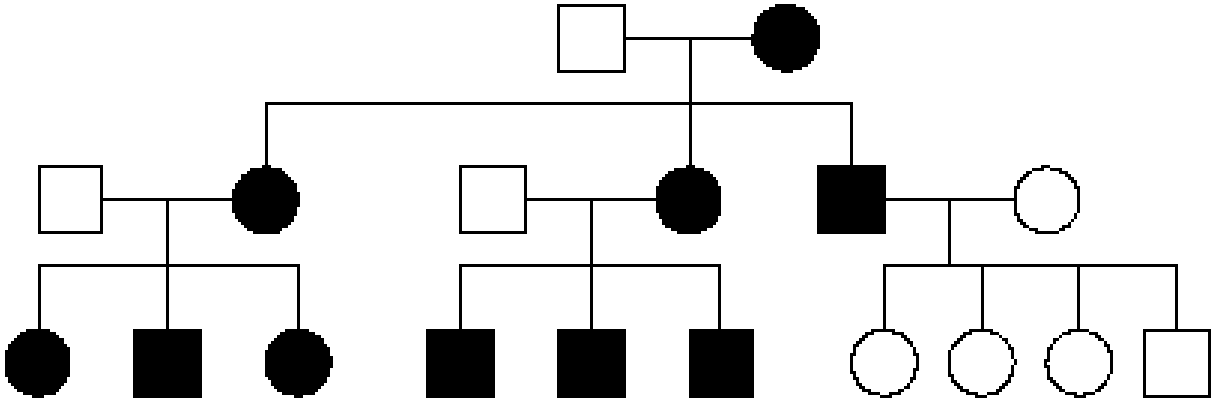
3. Colorblindness is a sex-linked recessive trait. A colorblind male and a female with normal vision have a son who is colorblind. What are the parents' genotypes?

Ch. 12 Warm-Up

1. What is a Barr body?
2. How are linkage maps constructed? (See. Fig. 12.11 in your textbook, *BIF 2e*).
3. Determine the sequence of genes along a chromosome based on the following recombination frequencies: A-B, 8 map units; A-C, 19 map units; A-D, 20 map units; B-C, 11 map units; B-D, 28 map units.
4. What does a frequency of recombination of 50% indicate?

Ch. 12 Warm-Up

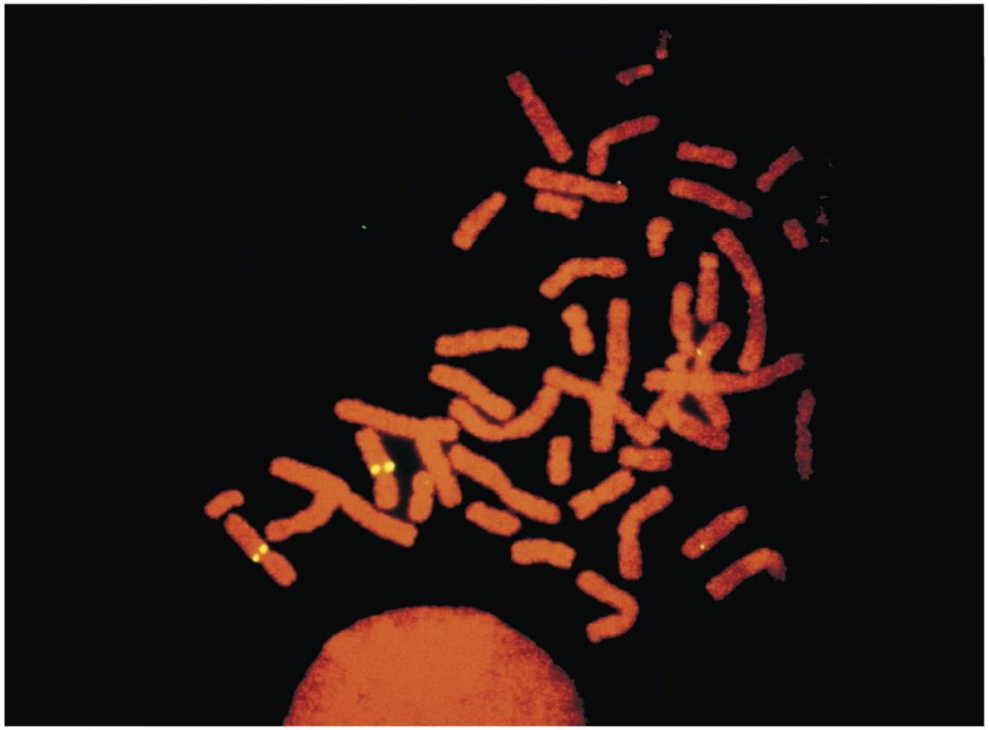
1. What is the pattern of inheritance of the trait (shaded square/circle) shown in the pedigree?

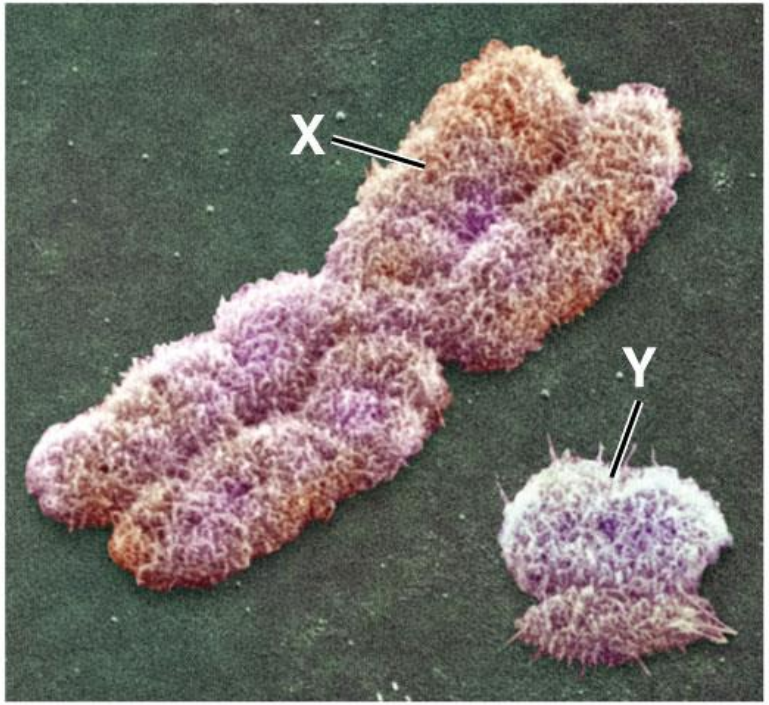


2. How many chromosomes are in a human cell that is:
   a) Diploid?
   b) Haploid?
   c) Triploid?
   d) Monosomic?
   e) Trisomic?

The Chromosomal Basis Of Inheritance

CHAPTER 12





What you must know:

- How the chromosome theory of inheritance connects the physical movement of chromosomes in meiosis to Mendel's laws of inheritance.
- The unique pattern of inheritance in sex-linked genes.
- How alteration of chromosome number or structurally altered chromosomes (deletions, duplications, etc.) can cause genetic disorders.
- How genetic imprinting and inheritance of mitochondrial DNA are exceptions to standard Mendelian inheritance.

Concept 12.1

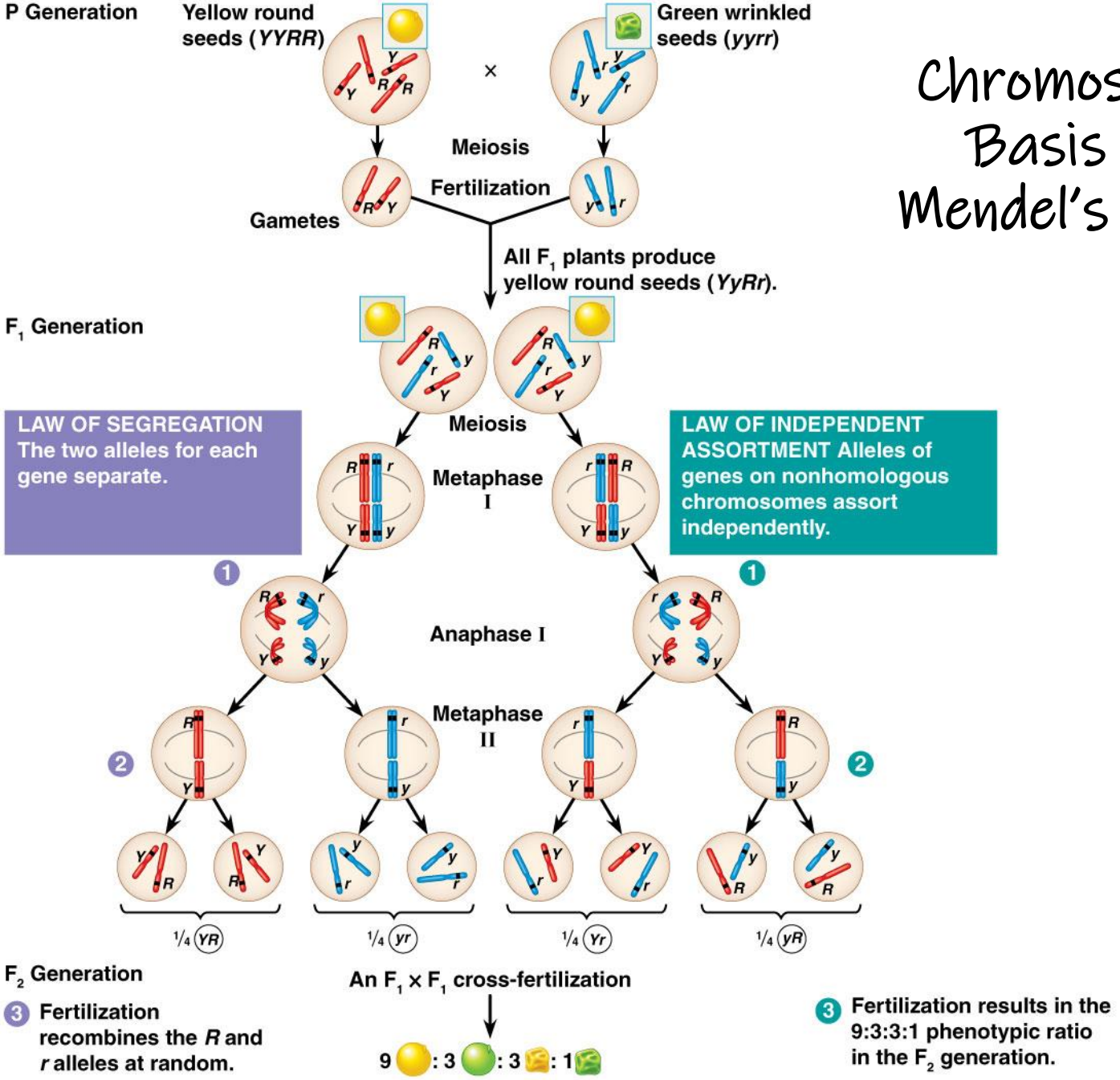
MORGAN SHOWED THAT MENDELIAN INHERITANCE HAS ITS PHYSICAL BASIS IN THE BEHAVIOR OF CHROMOSOMES

Chromosome Theory Of Inheritance

- Genes have specific loci (positions) along chromosomes
- Chromosomes undergo segregation and independent assortment

Chromosomes tagged to reveal a specific gene (yellow).

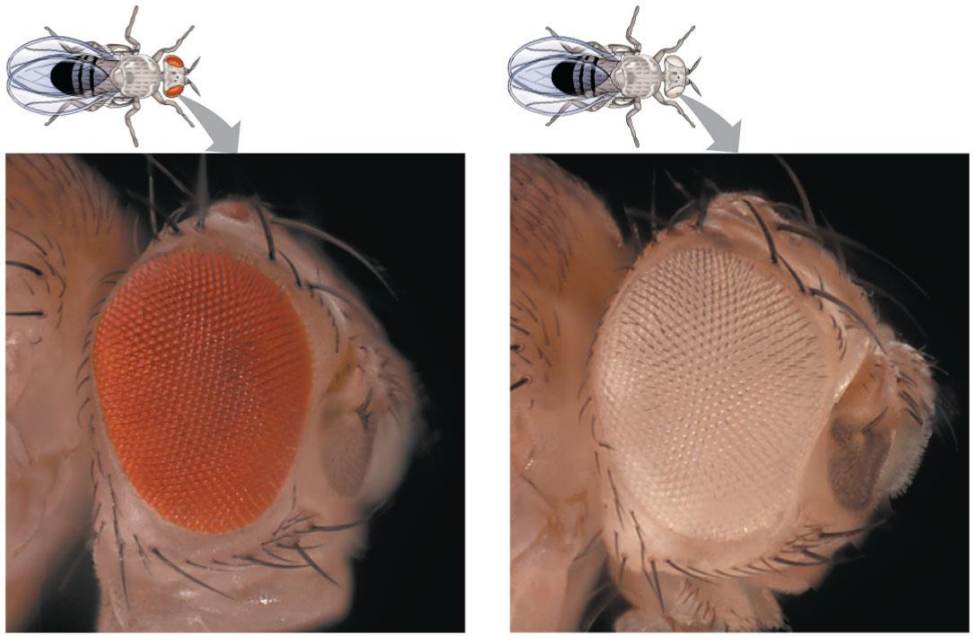
Chromosomal Basis Of Mendel's Laws

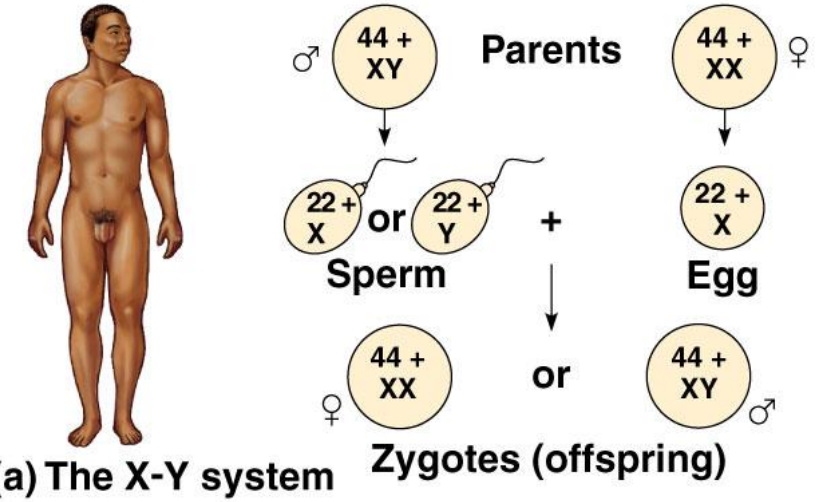


Thomas Hunt Morgan

- *Drosophila melanogaster* – fruit fly
  - Fast breeding, 4 prs. chromosomes (XX/XY)

- Sex-linked gene: located on X or Y chromosome
  - Red-eyes = wild-type; white-eyes = mutant
  - Specific gene carried on specific chromosome





(a) The X-Y system

(b) The X-0 system

(c) The Z-W system

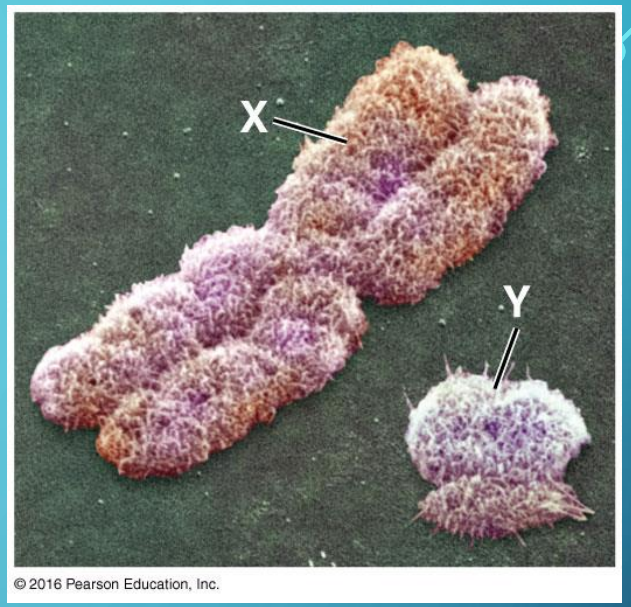
(d) The haplo-diploid system

Sex determination varies between animals

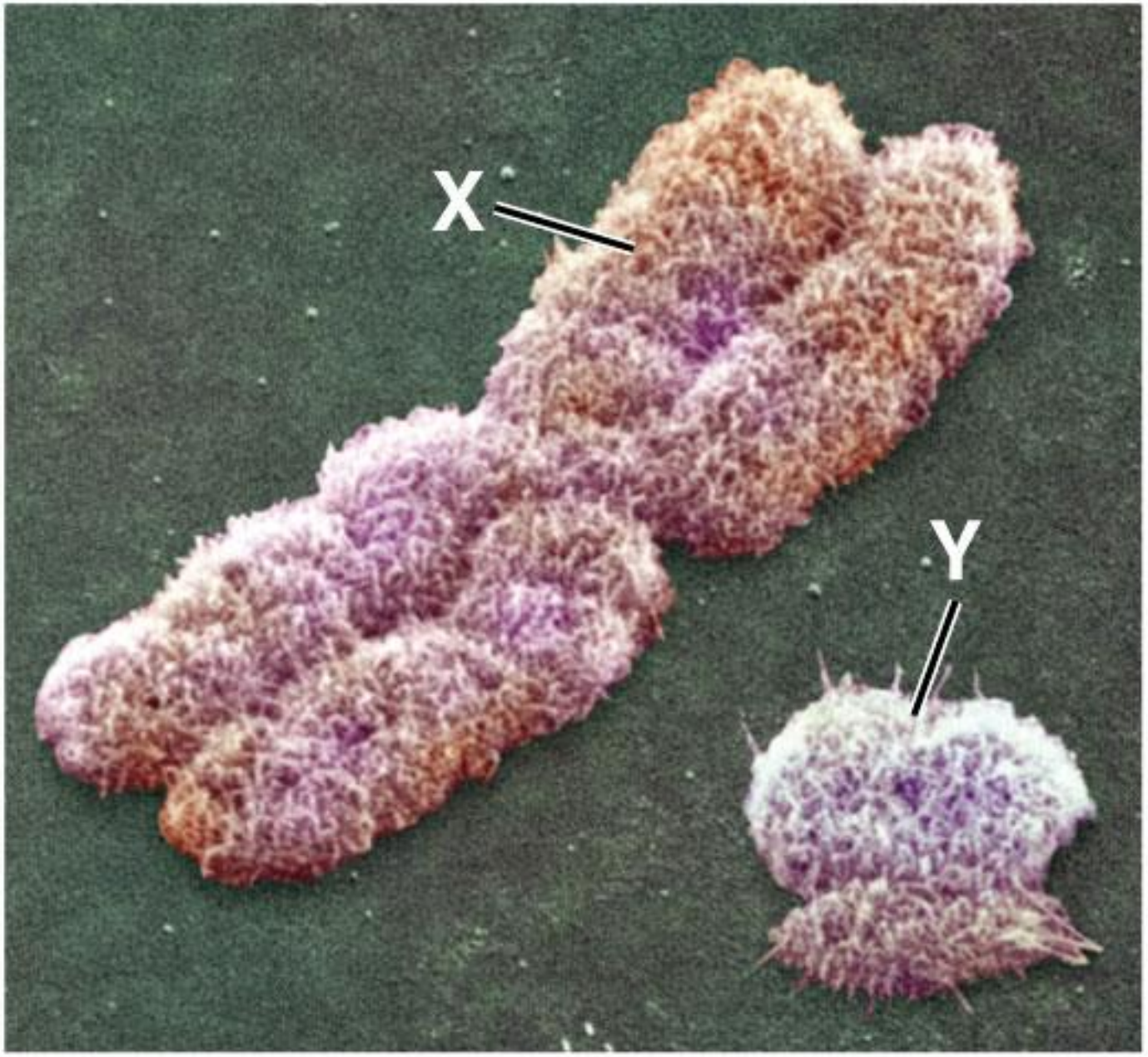
Concept 12.2

SEX-LINKED GENES EXHIBIT UNIQUE PATTERNS OF INHERITANCE

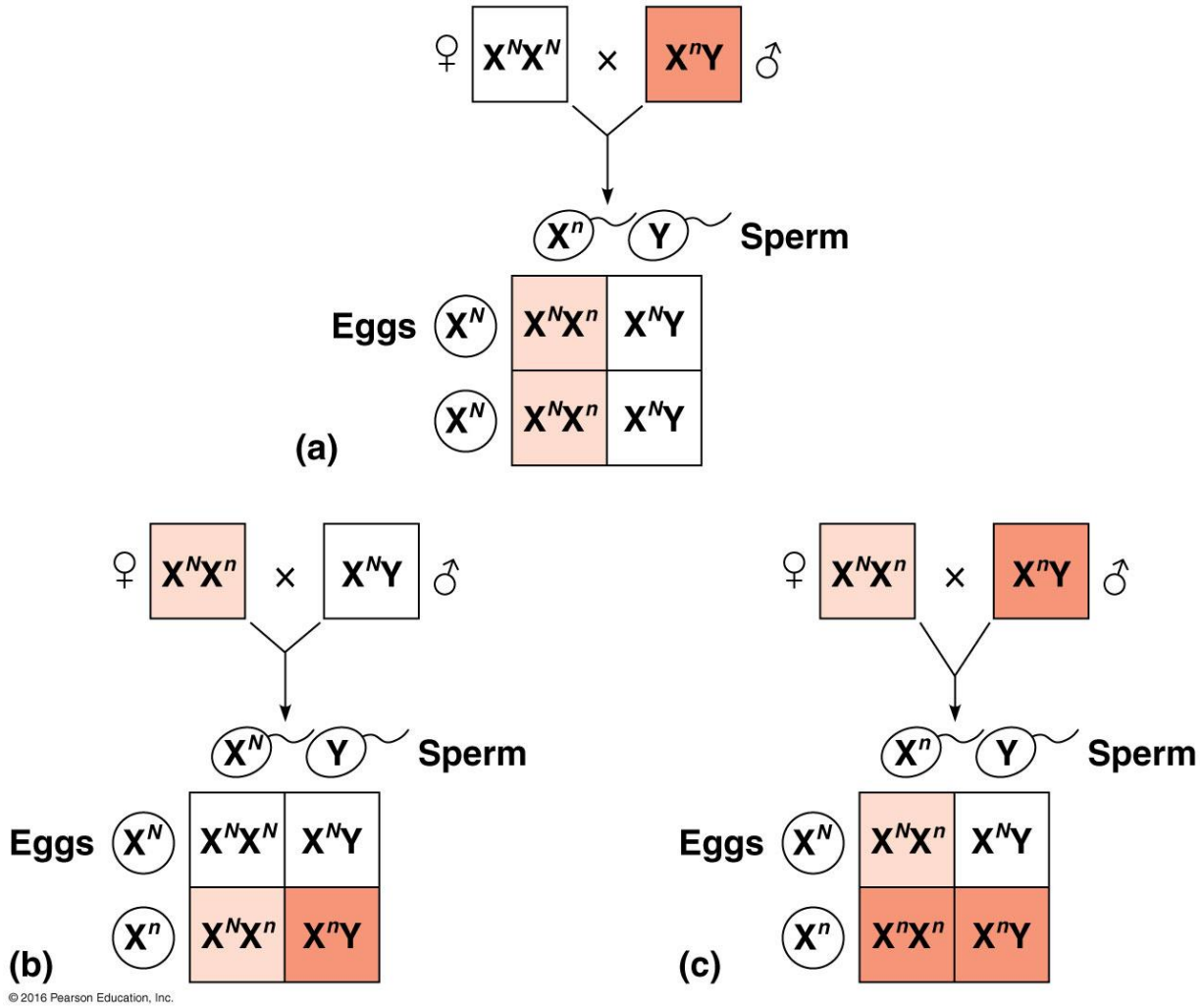
Sex-linked Genes



- Sex-linked gene on X or Y
- Females (XX), male (XY)
  - Eggs = X, sperm = X or Y
- Fathers pass X-linked genes to daughters, but not sons
- Males express recessive trait on the single X (*hemizygous*)
- Females can be affected or carrier



Transmission of X-linked recessive traits



Sex-linked Disorders

- Colorblindness
- Duchenne muscular dystrophy
- Hemophilia

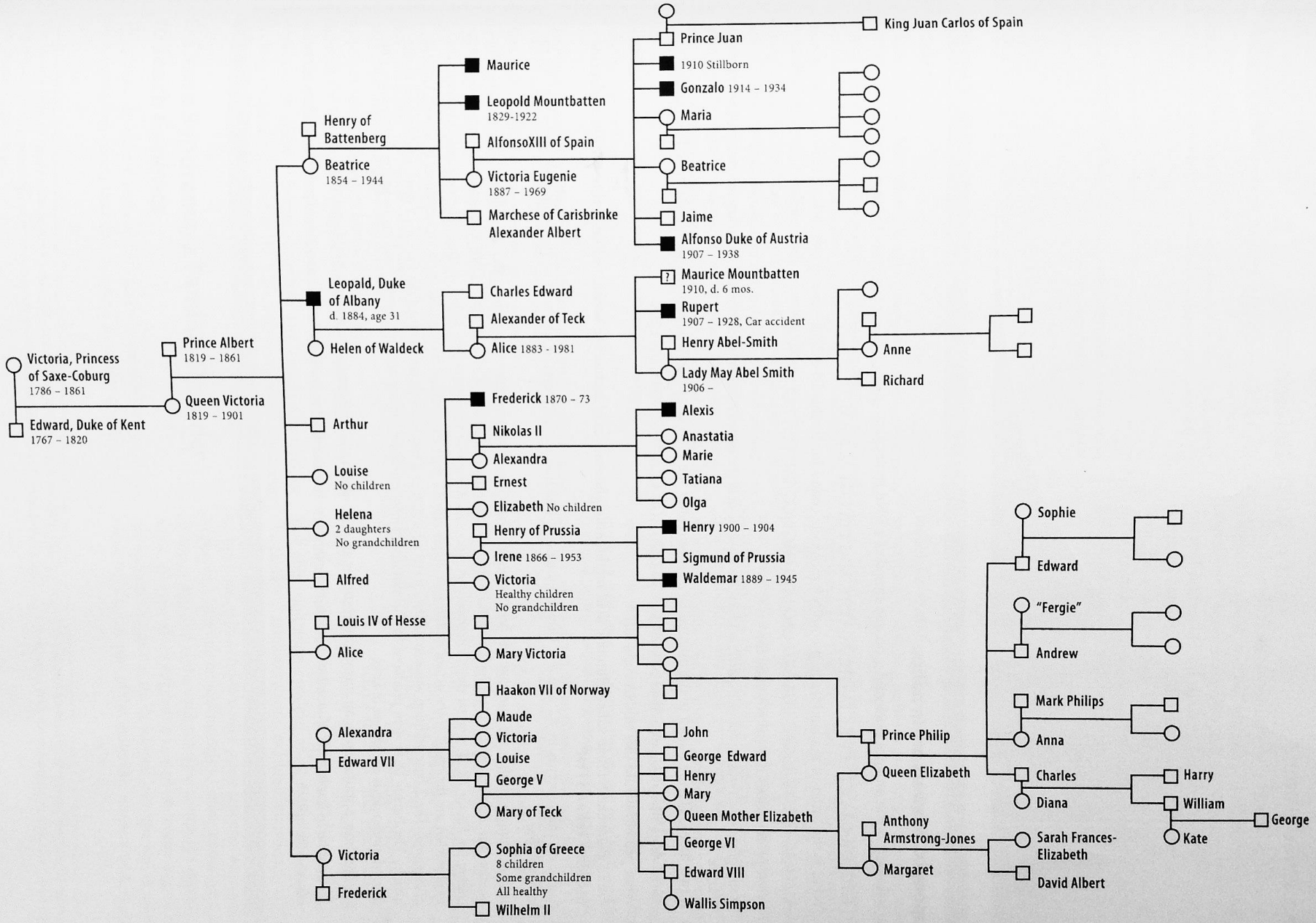


Figure 4. Partial pedigree of Queen Victoria and her descendants. Hemophiliac males are indicated by black squares.

X-Linked Genetics Practice Problem

A man with red-green colorblindness (a recessive, sex-linked condition) marries a woman with normal vision whose father was colour-blind. What is the probability that they will have a color-blind daughter? That their first son will be colour-blind?

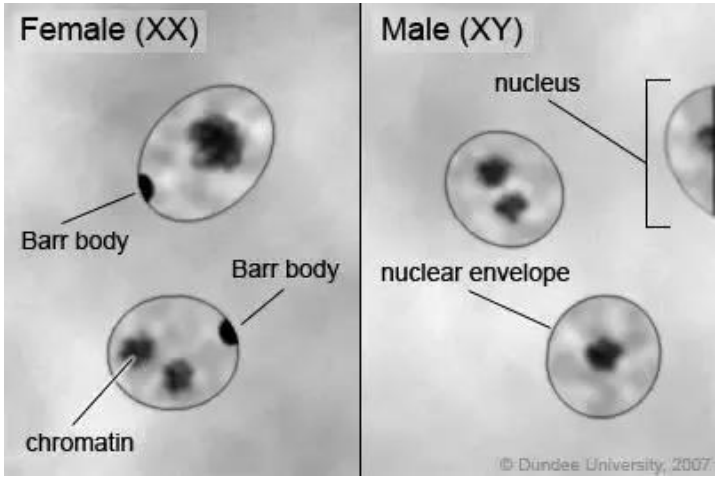
	X^b	Y
X^B	X^BX^b	X^BY
X^b	X^bX^b	X^bY

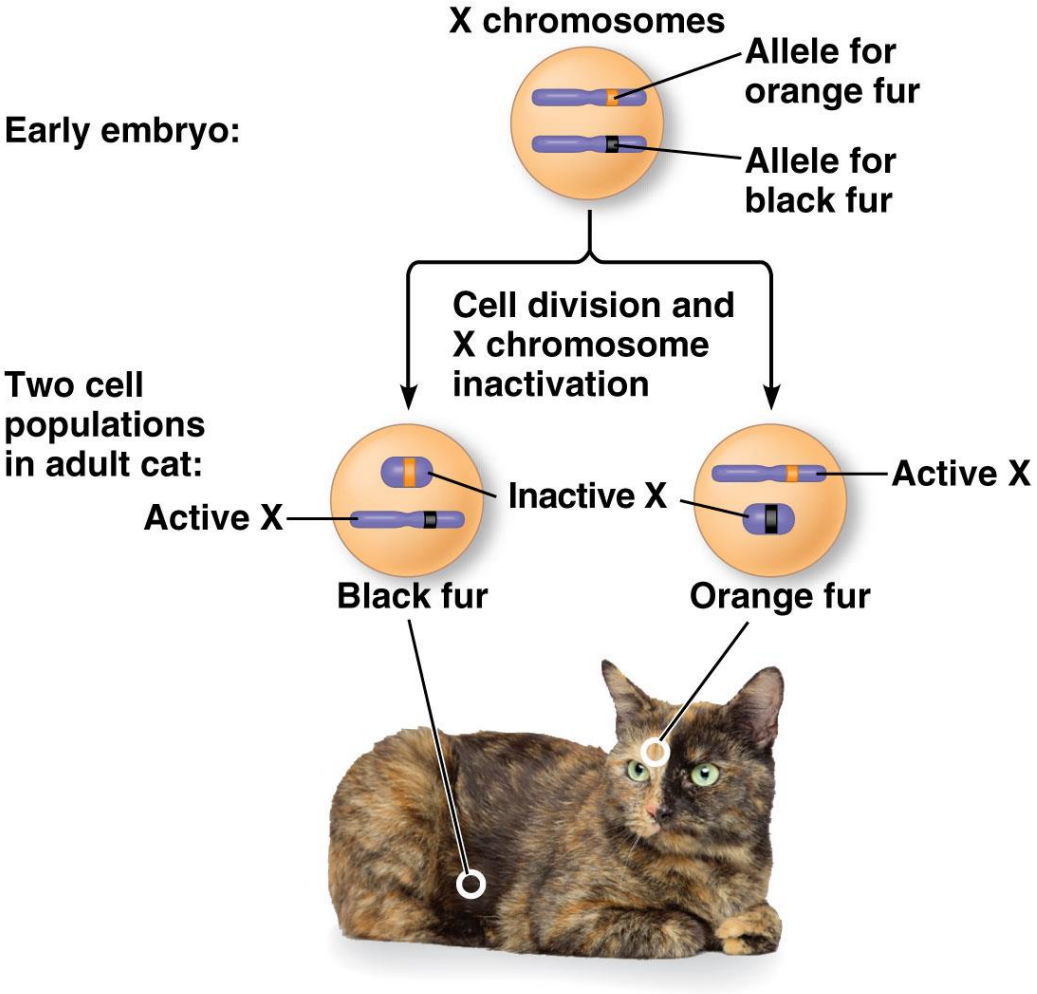
There's a 25% chance of a colour-blind daughter

Theres a 25% chance of a colour- blind son

X Inactivation

Barr body = inactive X chromosome; regulate gene dosage in females during embryonic development

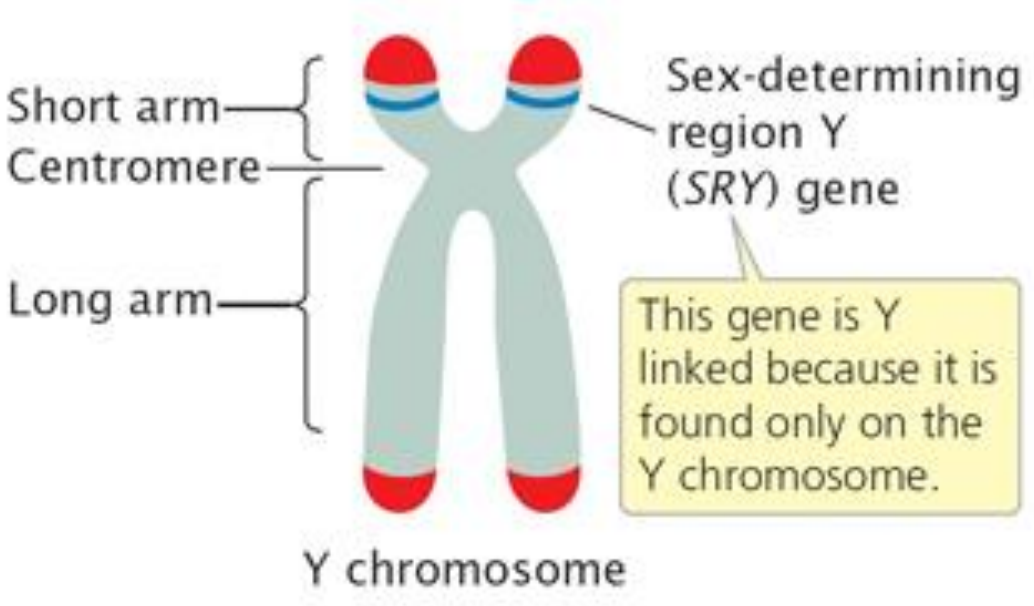




- Cats: allele for fur color is on X
- Only female cats can be tortoiseshell or calico.

Human Development

- Y chromosome required for development of testes
- Embryo gonads indifferent at 2 months
- SRY gene: sex-determining region of Y
- Codes for protein that regulates other genes

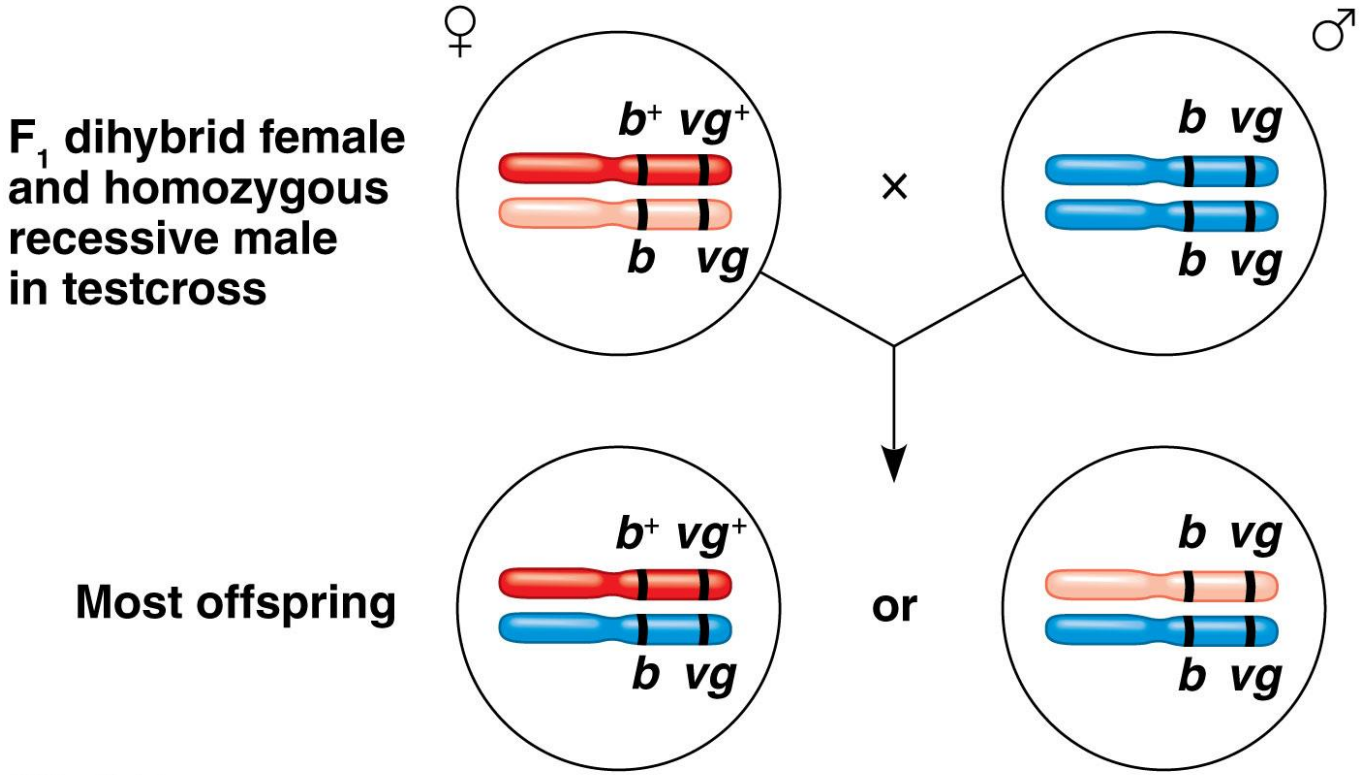


Concept 12.3

LINKED GENES TEND TO BE INHERITED TOGETHER BECAUSE THEY ARE LOCATED NEAR EACH OTHER ON THE SAME CHROMOSOME

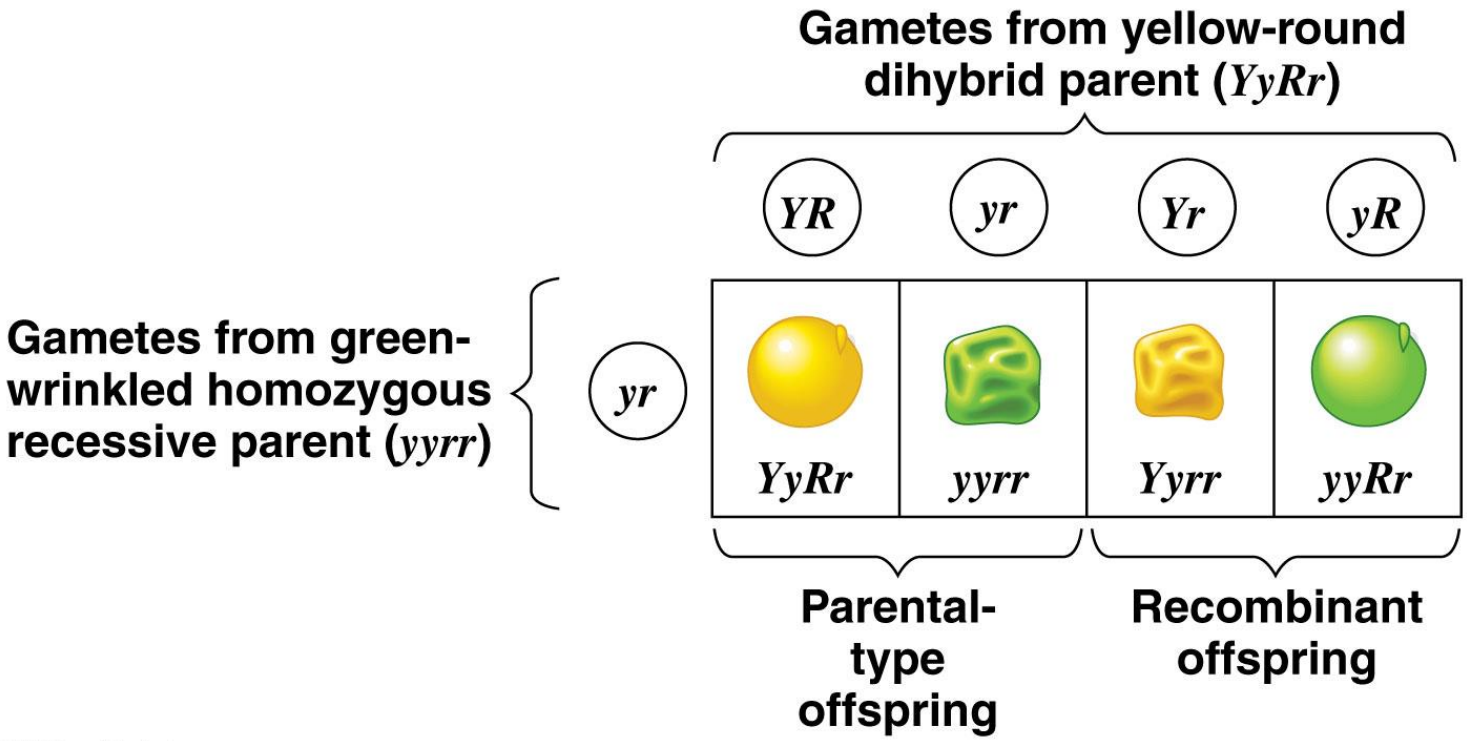
Linked Genes

- Genes located near each other on same chromosome tend to be inherited together



Genetic Recombination: production of offspring with combination of traits different from either parent

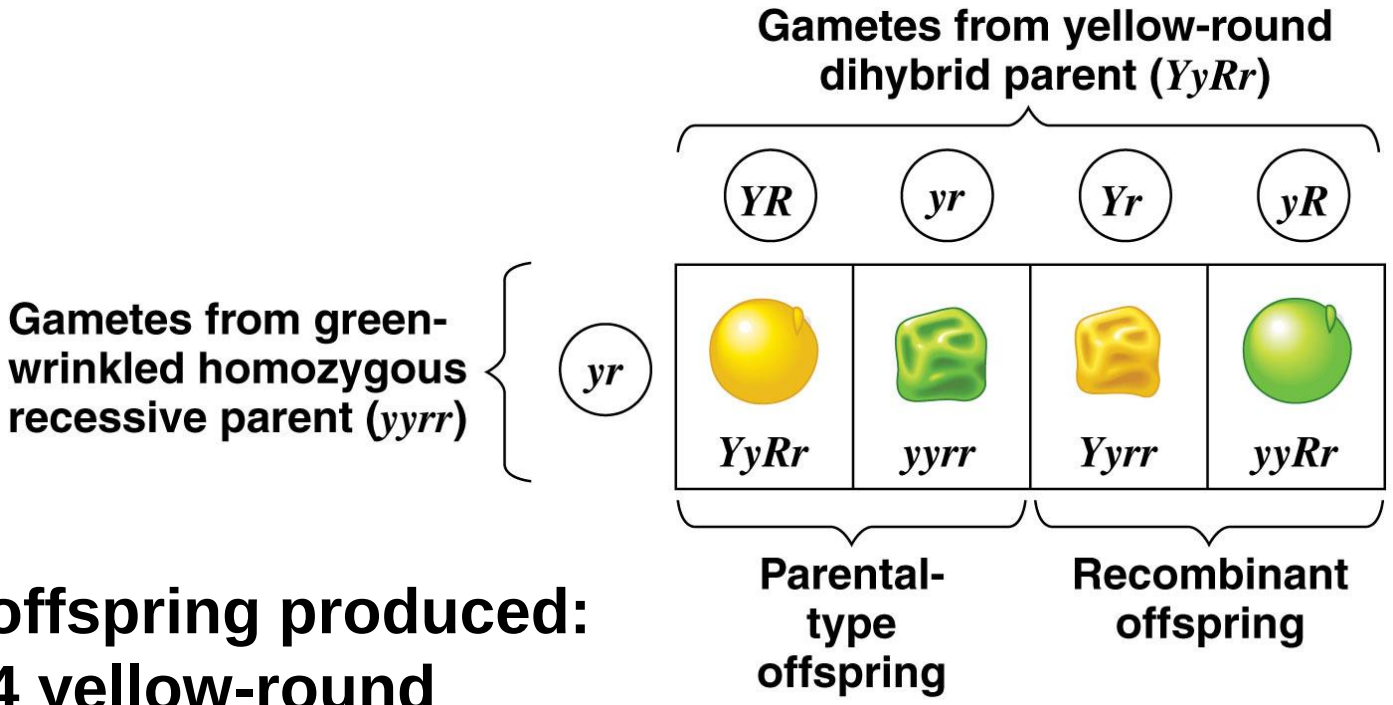
- If offspring look like parents → parental types
- If different from parents → recombinants



Calculating Recombination Frequency

$$\text{Recombination Frequency} = \frac{\text{\# Recombinants}}{\text{Total \# Offspring}} \times 100\%$$

Sample Problem 1: Calculate the recombination frequency

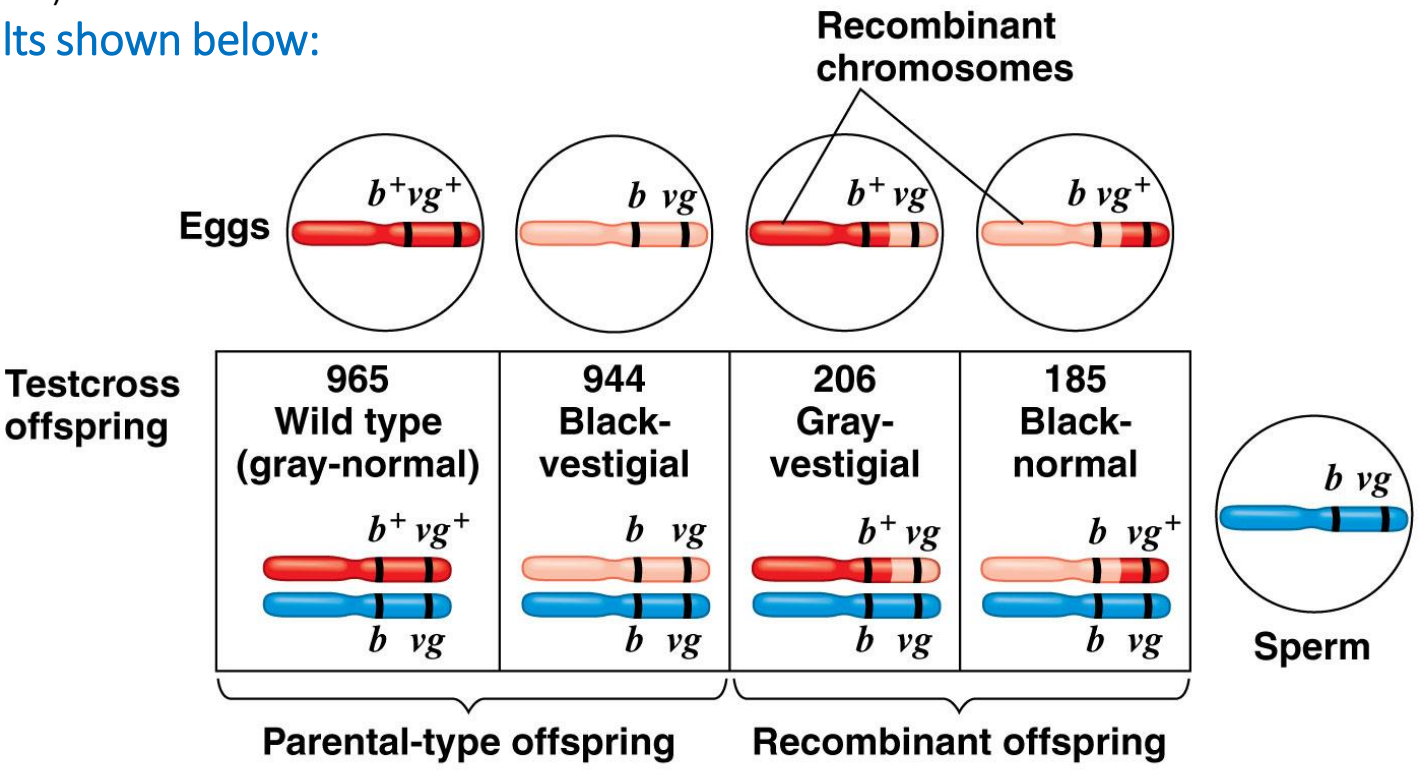


# of offspring produced:

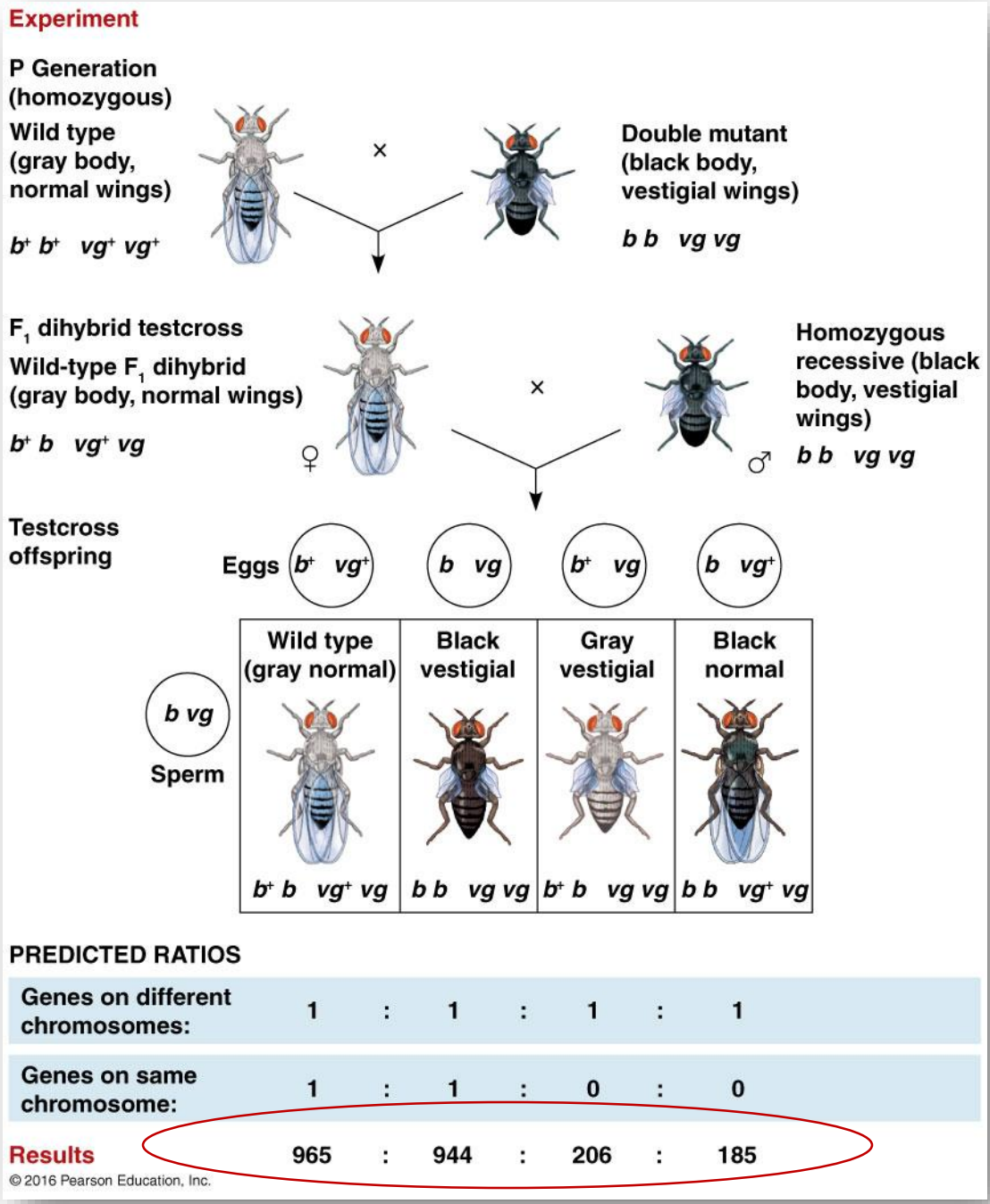
- **244 yellow-round**
- **256 green-wrinkled**
- **251 yellow-wrinkled**
- **249 green-round**

Sample Problem 2: Calculate the recombination frequency

- Original homozygous parents (P): Gray body, normal wings x black body, vestigial wings → F_1 dihybrid offspring
- Dyhybrid testcross (F_1): Gray, normal (heterzygous) x Black, vestigial (homozygous recessive)
- F_2 results shown below:

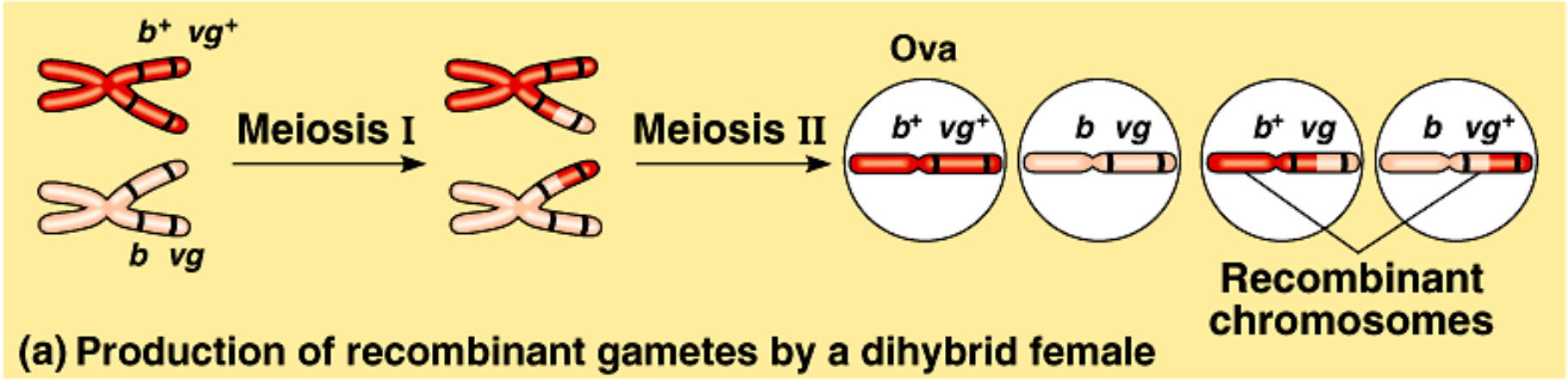


If results **do not** follow Mendel's Law of Independent Assortment, then **the genes are probably linked**

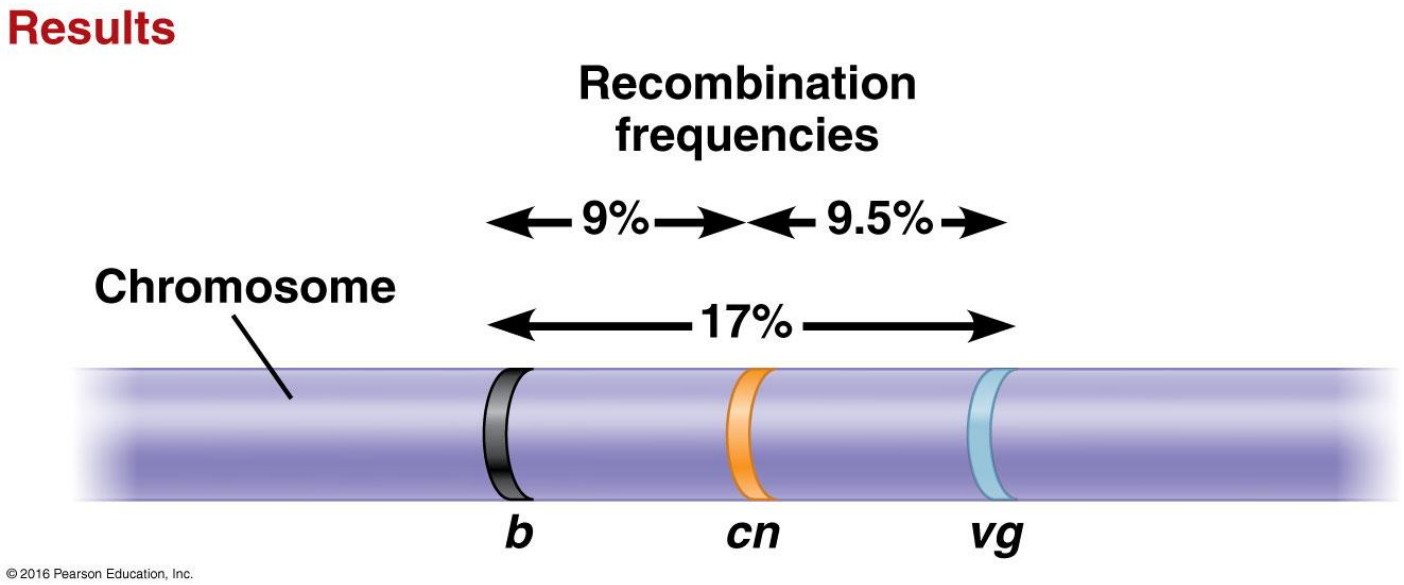


Crossing Over: explains why some linked genes get separated during meiosis

The further apart 2 genes on same chromosome, the higher the probability of crossing over and the higher the *recombination frequency*

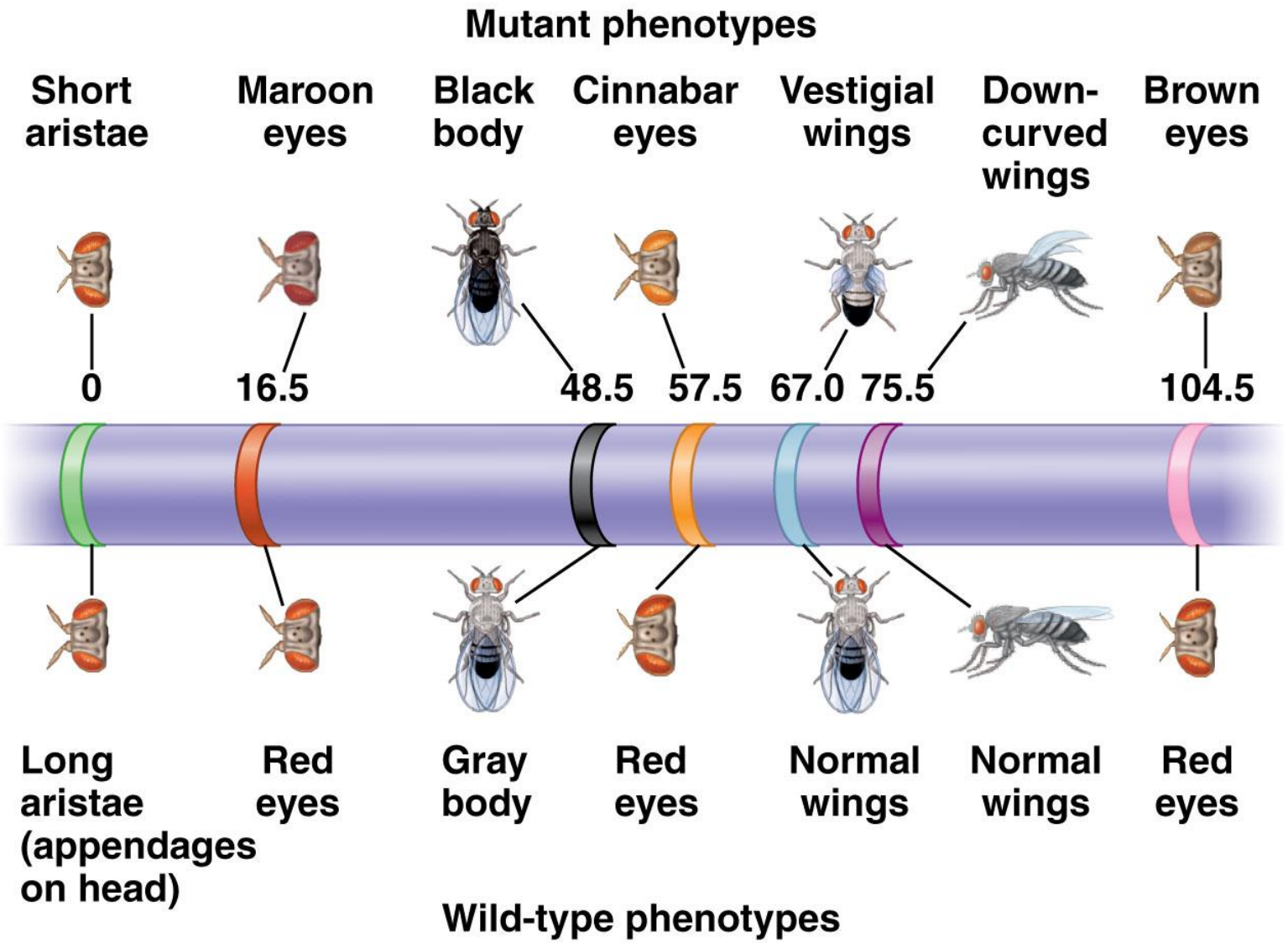


Linkage Map: genetic map that is based on % of crossover events



- 1 **map unit** = 1% recombination frequency
- Express relative distances along chromosome
- **50% recombination** = far apart on *same* chromosome or on 2 *different* chromosomes

Partial genetic (linkage) map of a *Drosophila* chromosome



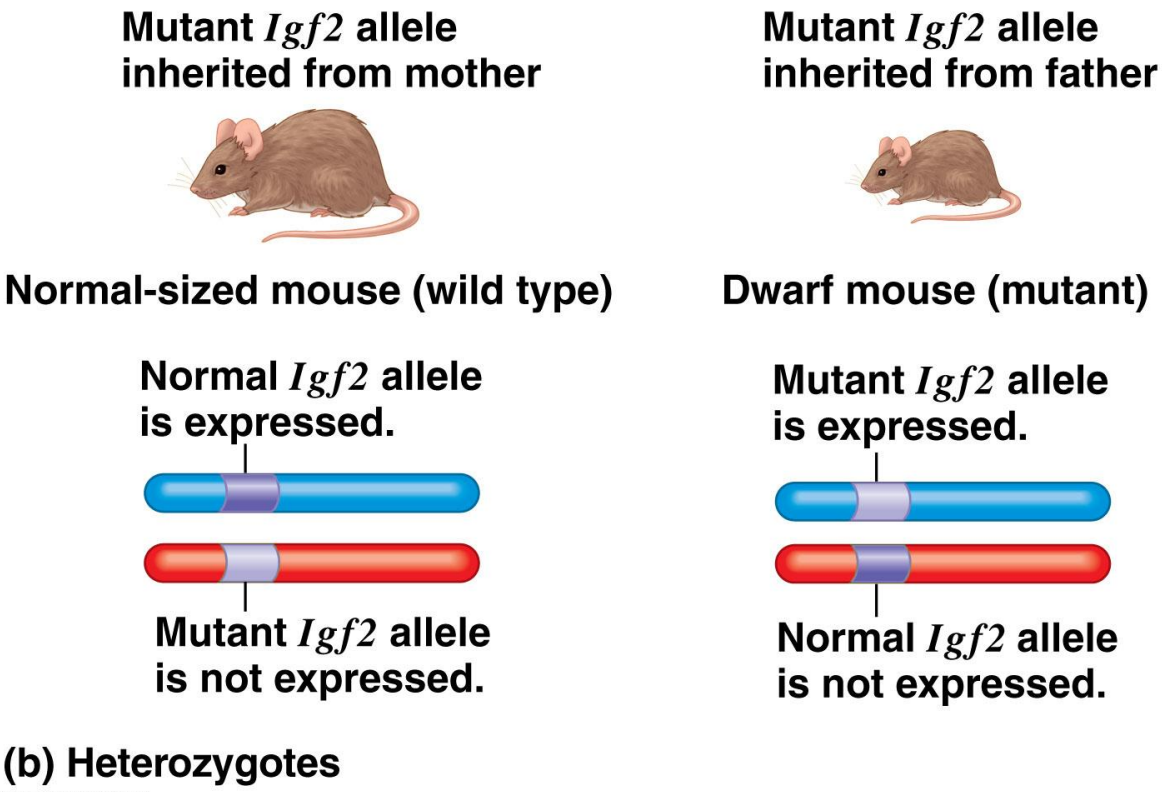
Linkage Map Sample Problem

Genes A, B and C are located on the same chromosome. Testcrosses show that the recombination frequency between A and B is 28% and between A and C is 12%. Can you determine the linear order of these genes? Explain.

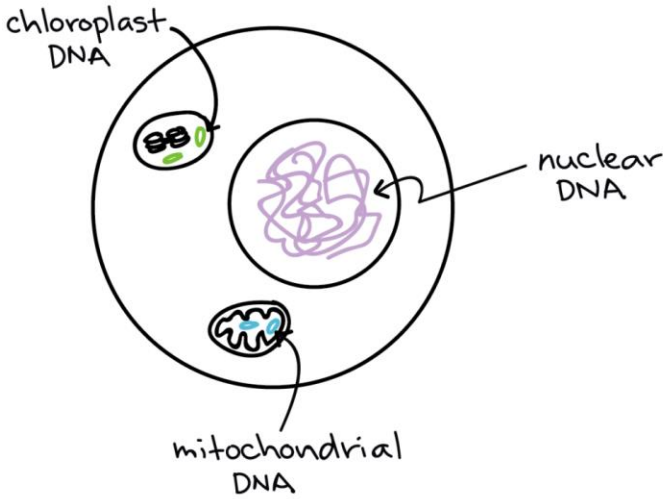
Exceptions To Mendelian Inheritance

Genomic Imprinting

- Genomic imprinting: phenotypic effect of gene depends on whether from M or F parent
- Methylation: silence genes by adding methyl groups to DNA



Non-Nuclear DNA

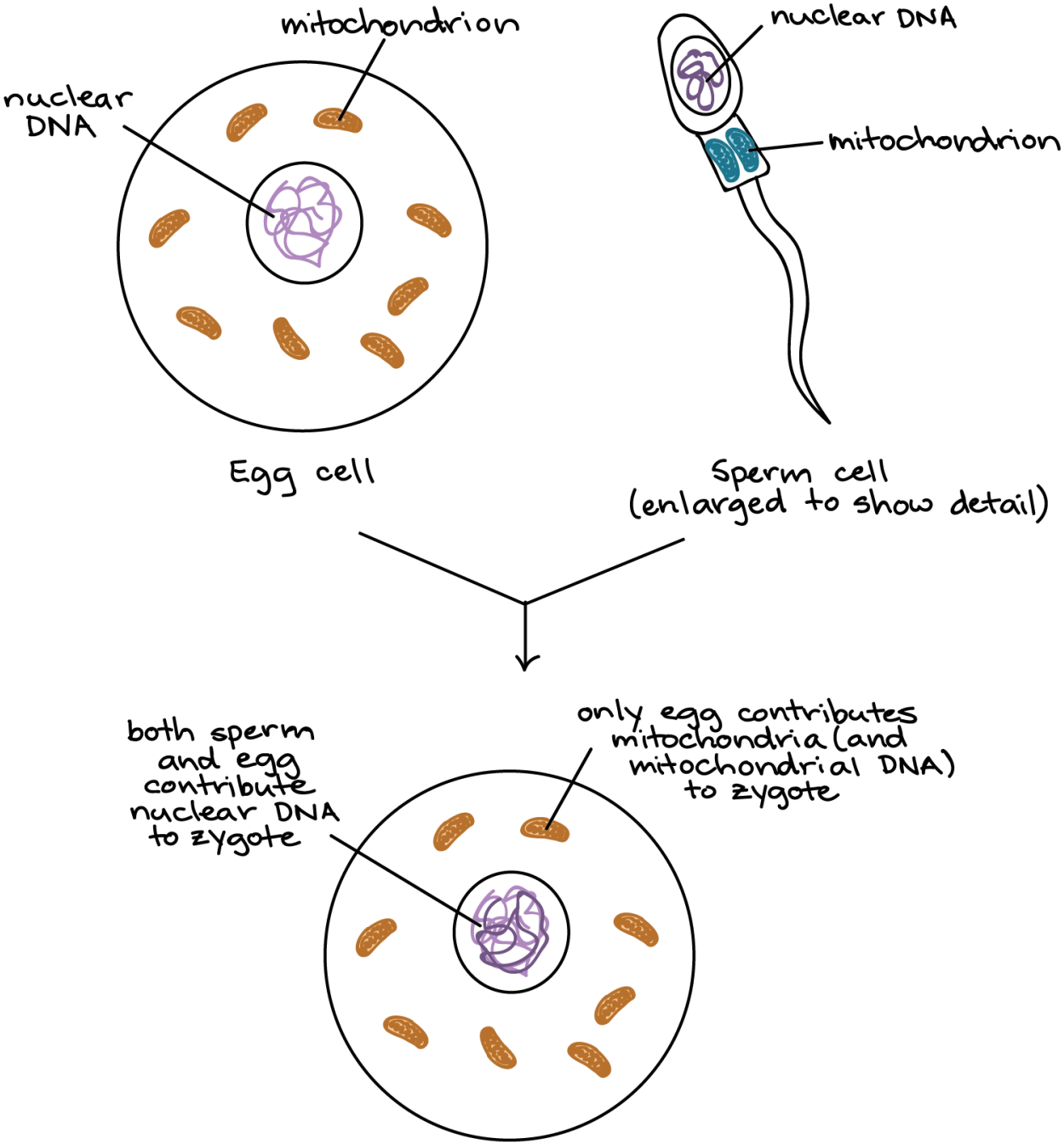


- Some genes located in **organelles**
  - Mitochondria, chloroplasts, plastids
  - Contain small circular DNA

Maternal Inheritance:

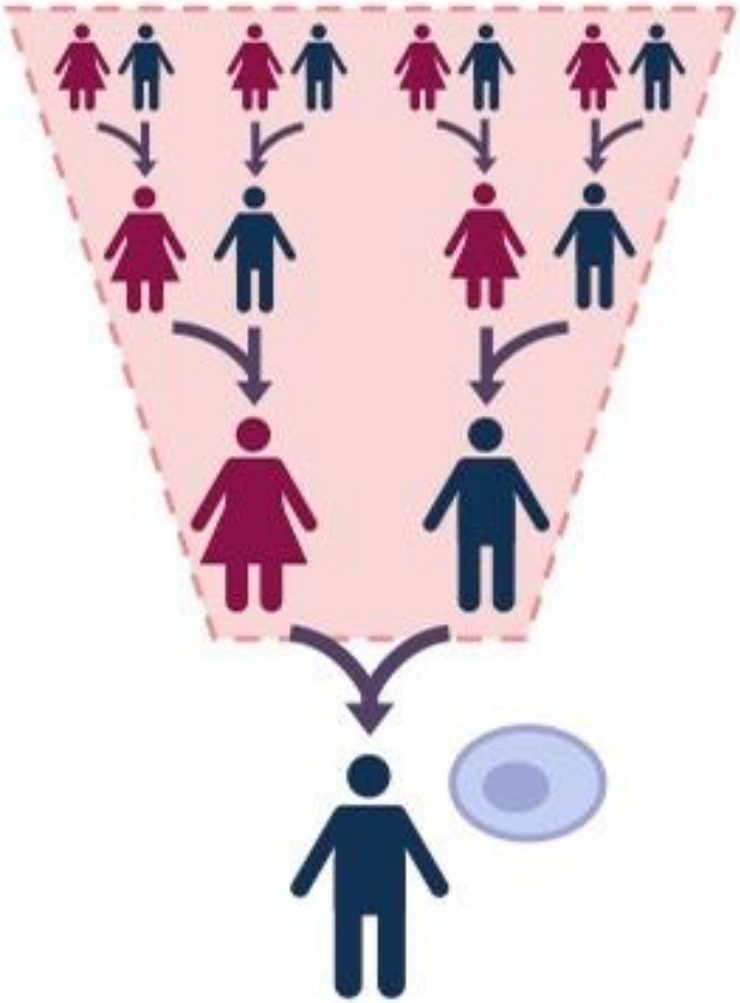
- In animals: mitochondria transmitted by the **egg** and not sperm
- In plants: mitochondria and chloroplasts transmitted in **ovule** and not pollen

Variegated (striped or spotted) leaves result from mutations in pigment genes in plastids, which generally are inherited from the maternal parent.


mitochondrion
nuclear DNA
nuclear DNA
mitochondrion
Egg cell
Sperm cell
(enlarged to show detail)
both sperm and egg contribute nuclear DNA to zygote
only egg contributes mitochondria (and mitochondrial DNA) to zygote

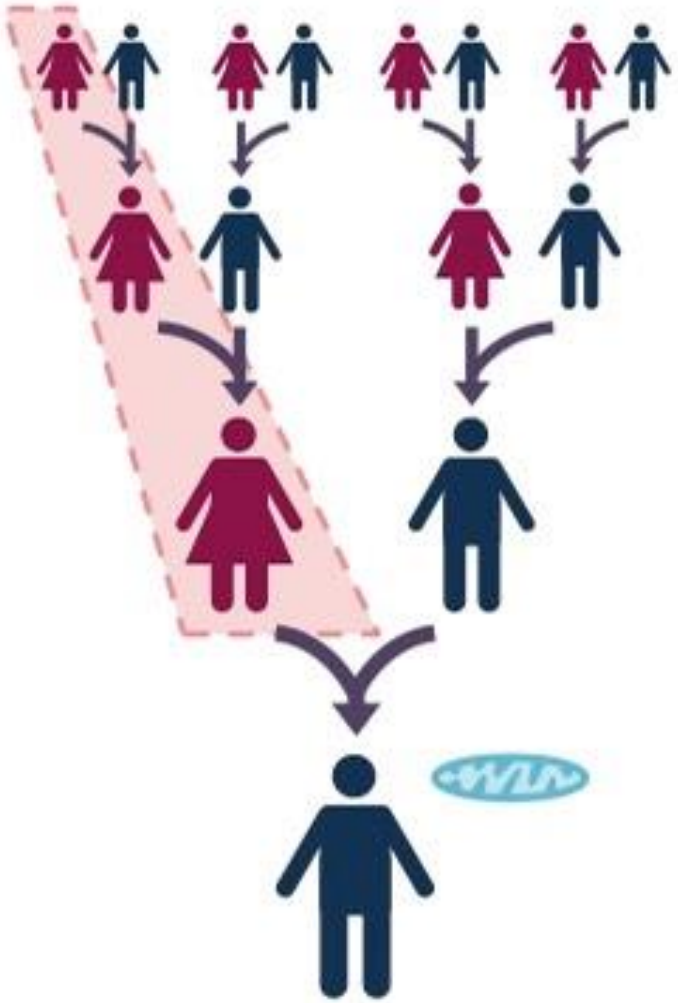
Nuclear DNA

Inherited from **all** ancestors



Mitochondrial DNA (mtDNA)

Inherited from a **maternal** lineage



Concept 12.4

ALTERATIONS OF CHROMOSOME NUMBER OF STRUCTURE CAUSE SOME GENETIC DISORDERS

Genetic Testing

Reasons for Genetic Tests:

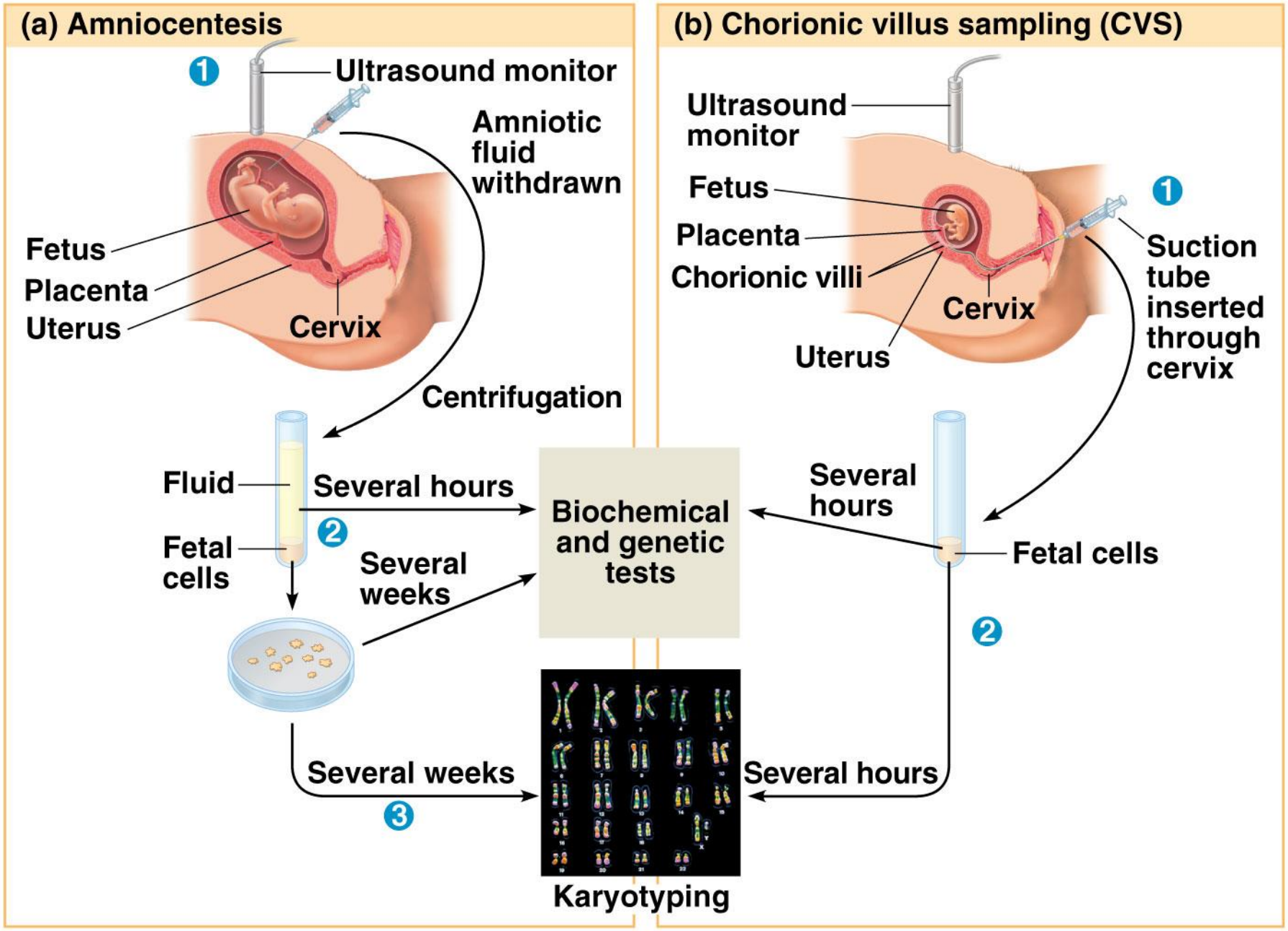
- Diagnostic testing (genetic disorders)
- Presymptomatic & predictive testing
- Carrier testing (before having children)
- Pharmacogenetics (medication & dosage)
- Prenatal testing
- Newborn screening
- Preimplantation testing (embryos)

Prenatal Testing

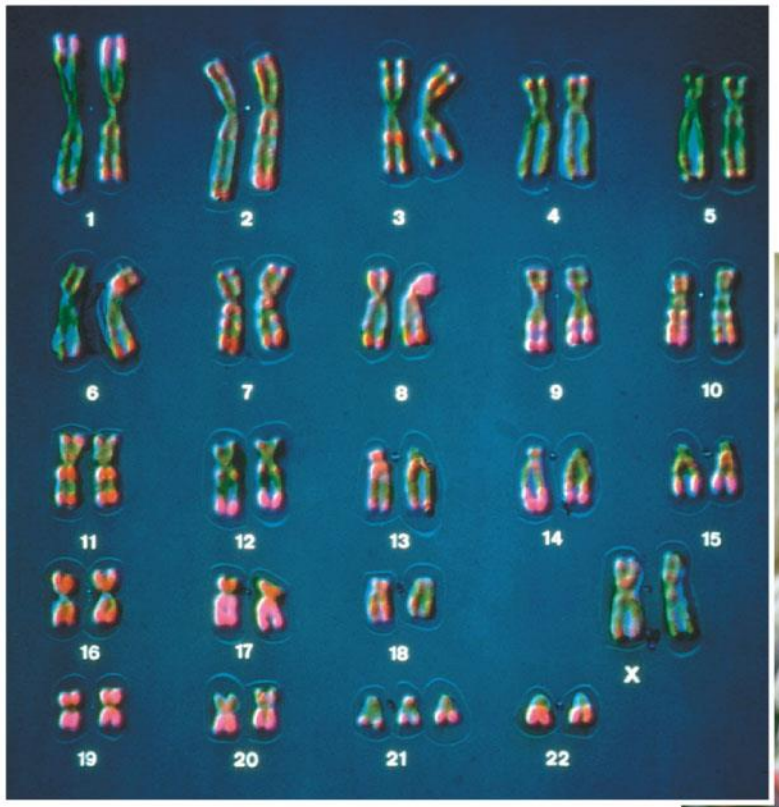
- May be used on a fetus to detect genetic disorders

- Amniocentesis: remove amniotic fluid around fetus to culture for karyotype

- Chorionic villus sampling: insert narrow tube in cervix to extract sample of placenta with fetal cells for karyotype

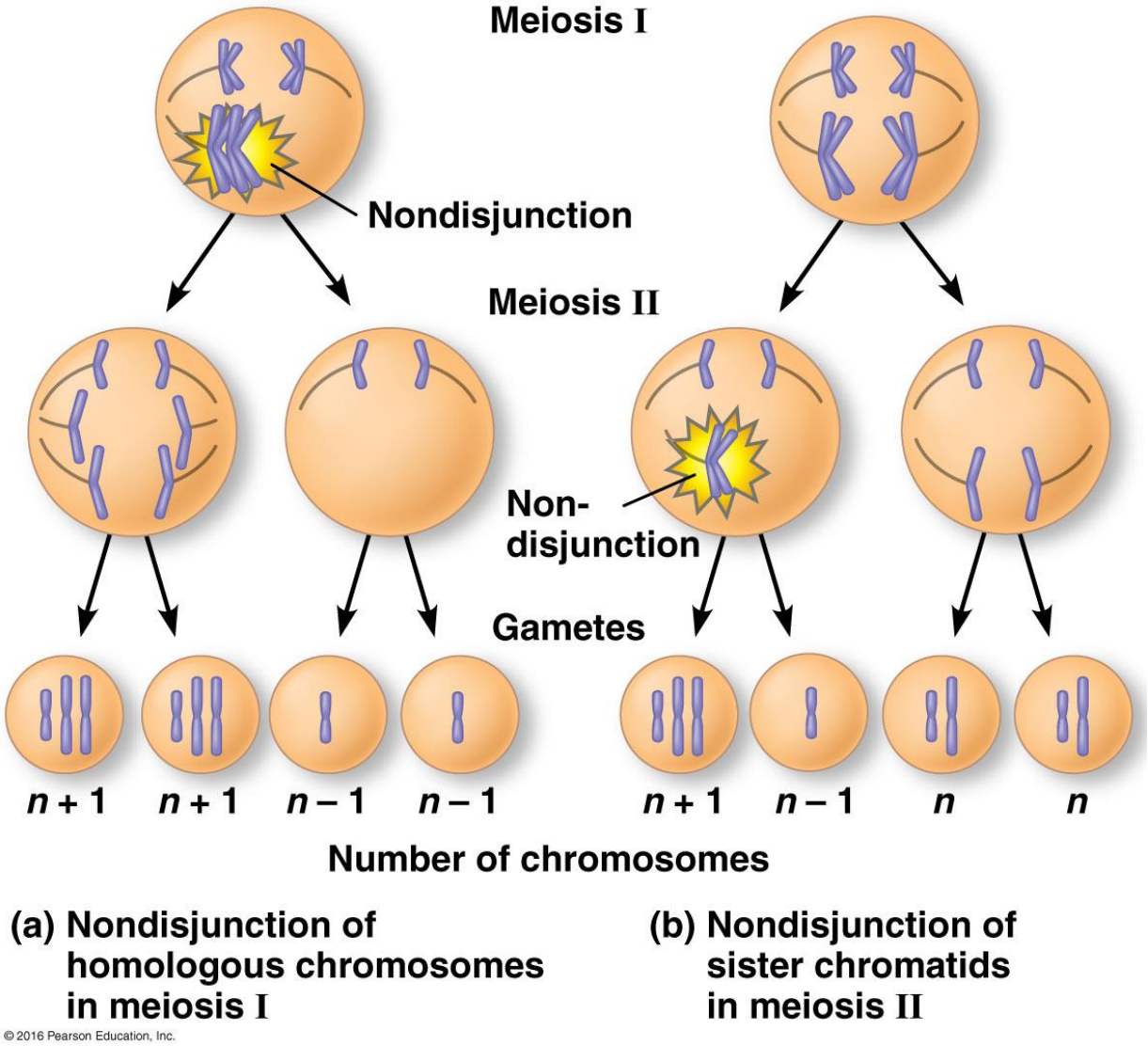


Karyotyping can detect nondisjunctions.



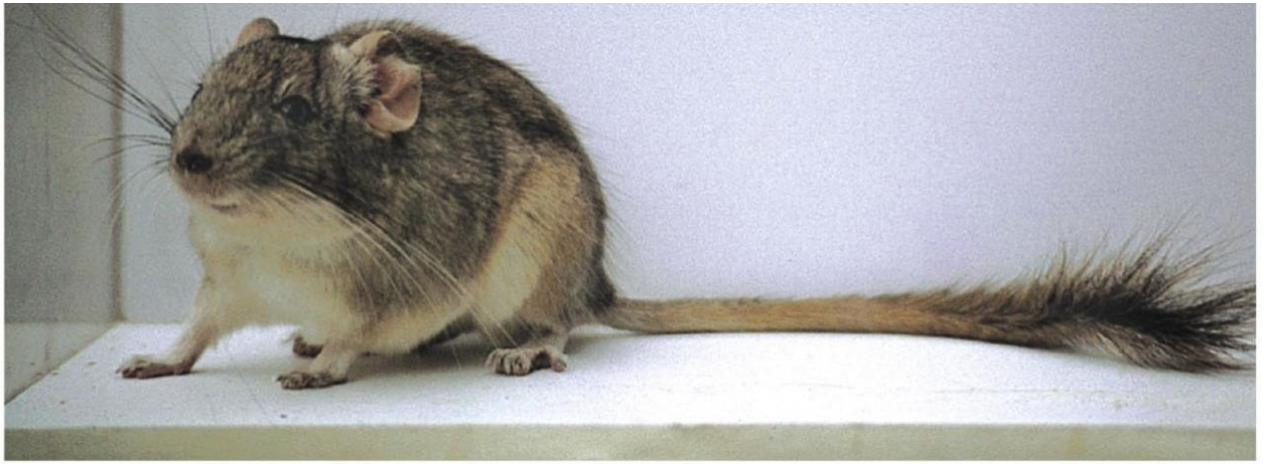
Down Syndrome = Trisomy 21

Nondisjunction: chromosomes fail to separate properly in Meiosis I or Meiosis II



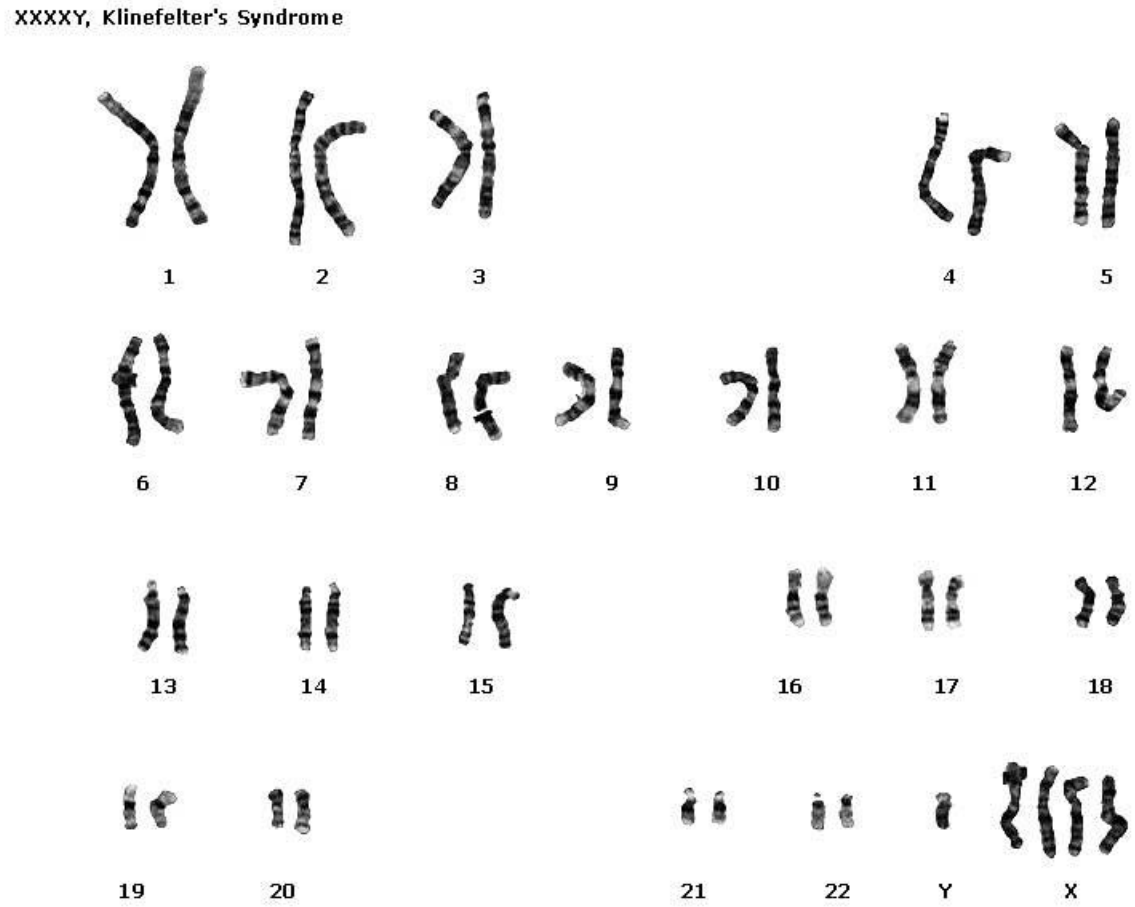
Nondisjunction

- Aneuploidy: abnormal # chromosomes
  - Monosomic (1 copy → 2n-1)
  - Trisomic (3 copies → 2n+1)
- Polyploidy: 2+ complete sets of chromosomes
  - triploid (3n) or tetraploid (4n)
  - rare in animals, frequent in plants (wheat, strawberries)



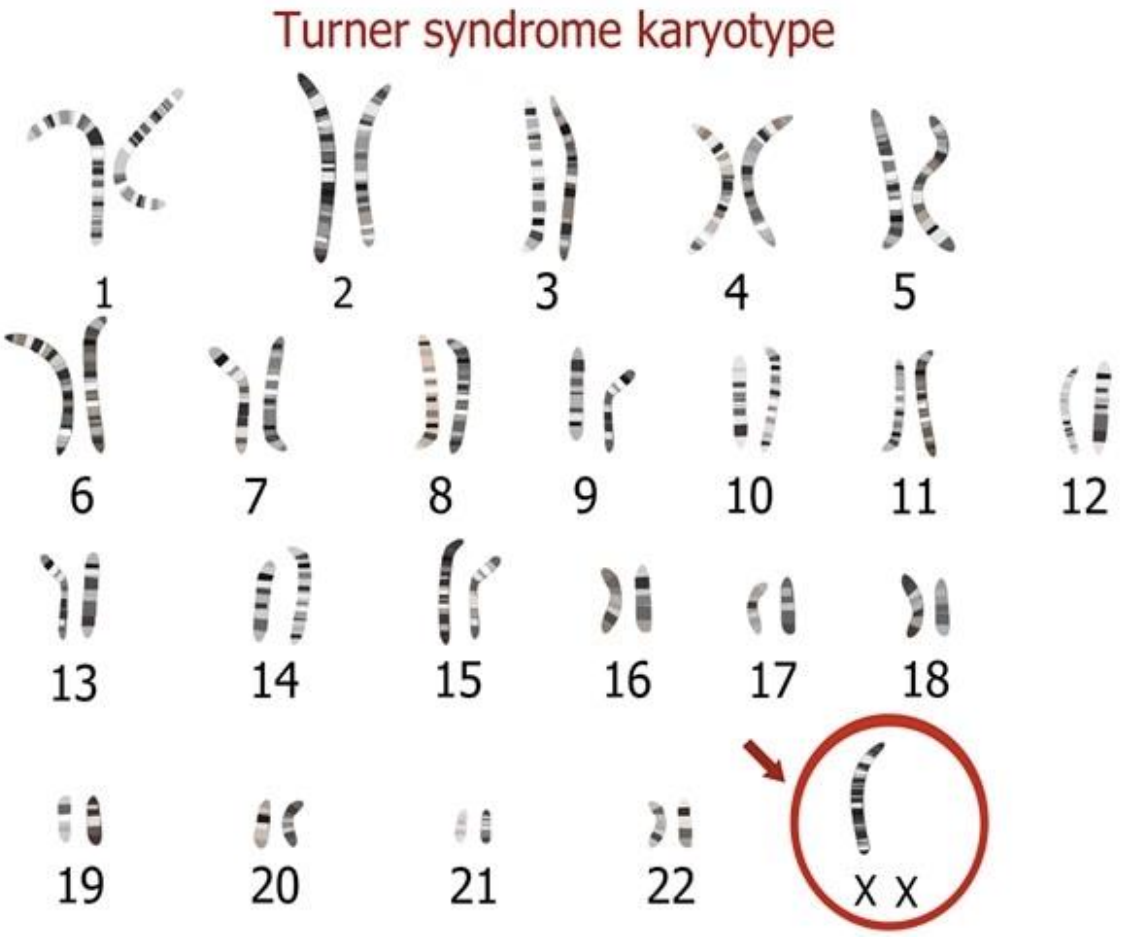
A tetraploid mammal. Scientists think this species may have arisen when an ancestor doubled its chromosome # by errors in mitosis or meiosis.

Nondisjunction



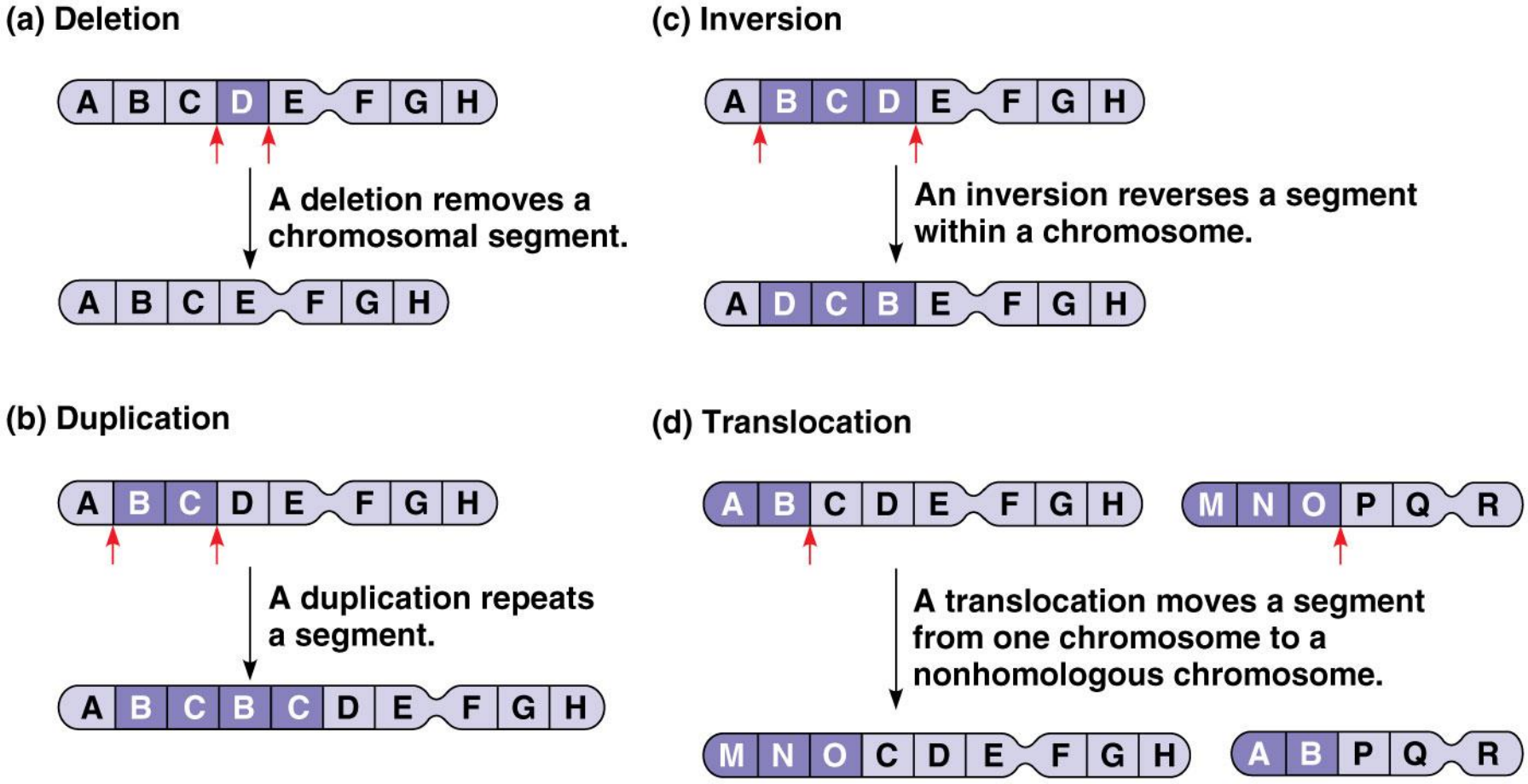
Klinefelter Syndrome: 47XYY, 47XXY

Nondisjunction

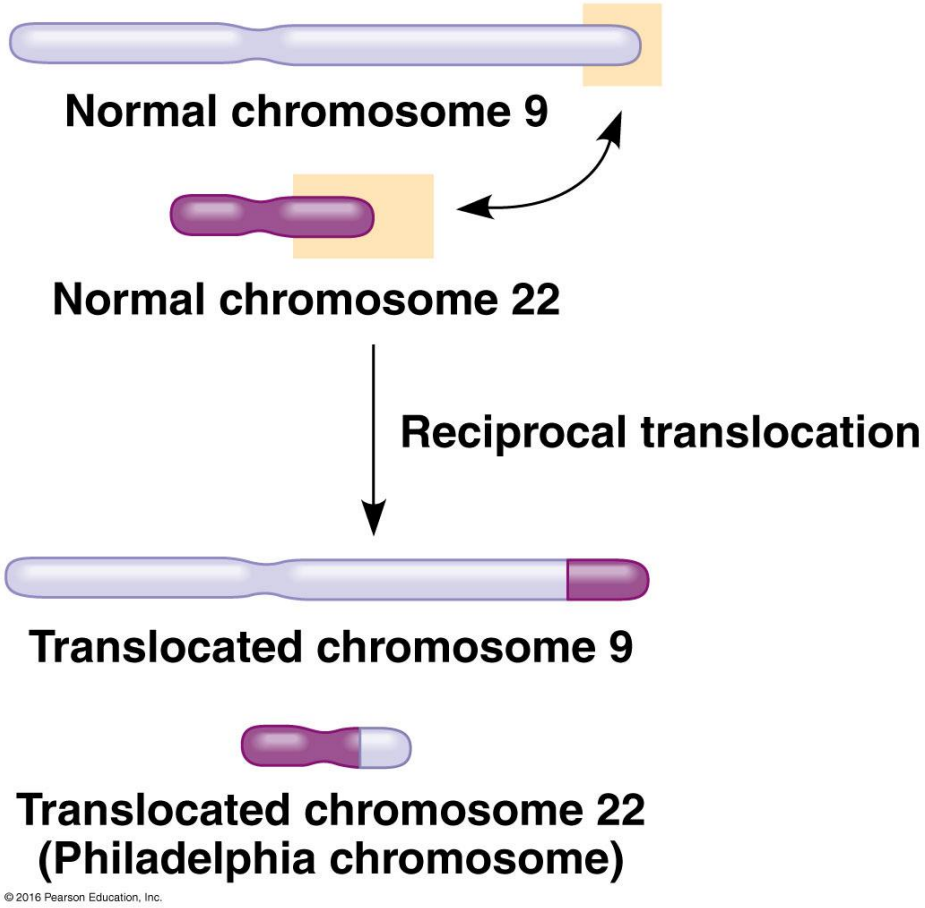


Turner Syndrome = 45XO

Chromosomal Mutations

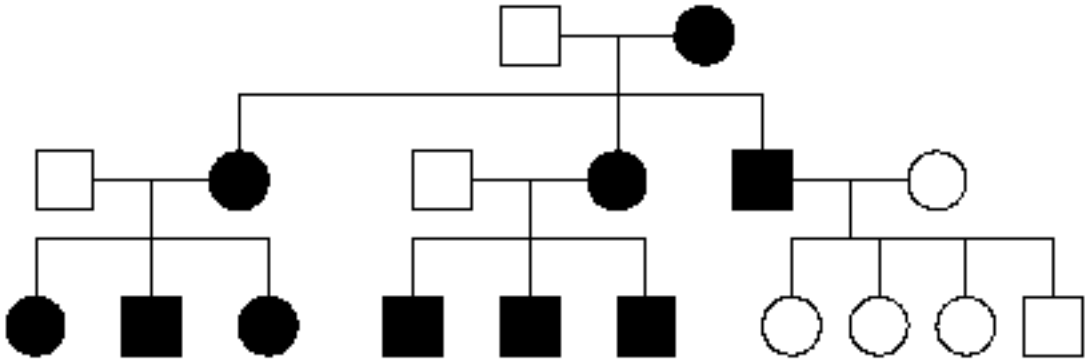


Translocation associated with chronic myelogenous leukemia (CML) → cancer



Review Questions

1. What is the pattern of inheritance of the trait (shaded square/circle) shown in the pedigree?



2. How many chromosomes are in a human cell that is:

   a) Diploid?
   b) Triploid?
   c) Monosomic?
   d) Trisomic?

Chi-Squared Analysis Practice

Two true-breeding Drosophila are crossed: a normal-winged, red-eyed female and a miniature-winged, vermillion-eyed male. The F_1 offspring all have normal wings and red eyes. When the F_1 offspring are crossed with miniature-winged, vermillion-eyed flies, the following offspring resulted:

- 233 normal wing, red eye
- 247 miniature wing, vermillion eye
- 7 normal wing, vermillion eye
- 13 miniature wing, red eye

What types of conclusions can you draw from this experiment? Explain your answer.