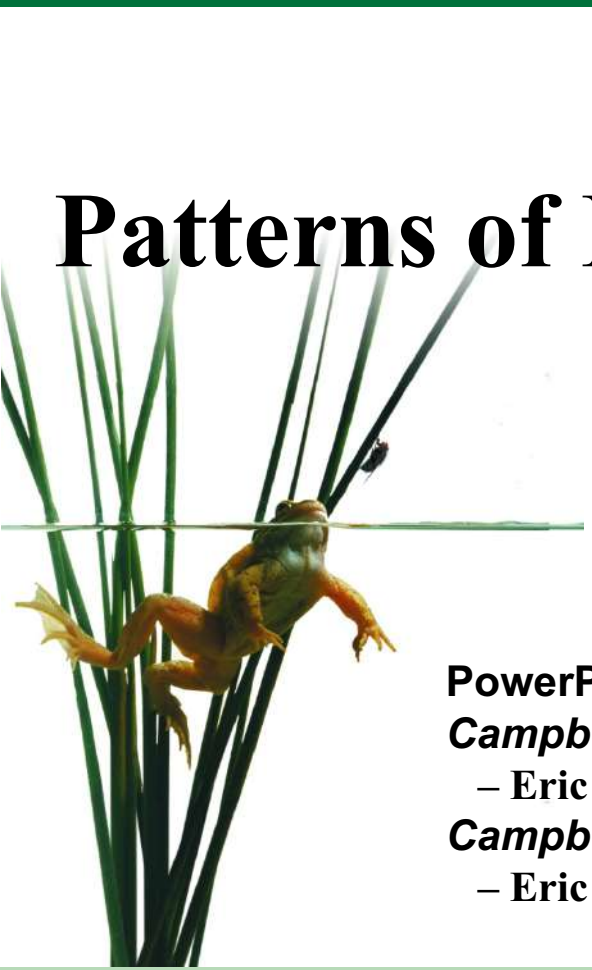
Chapter 9

Patterns of Inheritance



PowerPoint® Lectures for
***Campbell Essential Biology,* Fourth Edition**
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***Campbell Essential Biology with Physiology,* Third Edition**
– Eric Simon, Jane Reece, and Jean Dickey

Lectures by Chris C. Romero, updated by Edward J. Zalisko

Biology And Society: A Matter of Breeding

- **Genetics** is the scientific study of heredity.
  - Genetics explains why the offspring of purebred dogs are like their parents.
  - Inbreeding of dogs makes some genetic disorders common.
- A dog's behavior is determined by its
  - Genes
  - Environment

HERITABLE VARIATION AND PATTERNS OF INHERITANCE

- **Heredity** is the transmission of traits from one generation to the next.
- Gregor Mendel
  - Worked in the 1860s
  - Was the first person to analyze patterns of inheritance
  - Deduced the fundamental principles of genetics

In an Abbey Garden

- Mendel studied garden peas because they
  - Are easy to grow
  - Come in many readily distinguishable varieties
  - Are easily manipulated
  - Can self-fertilize

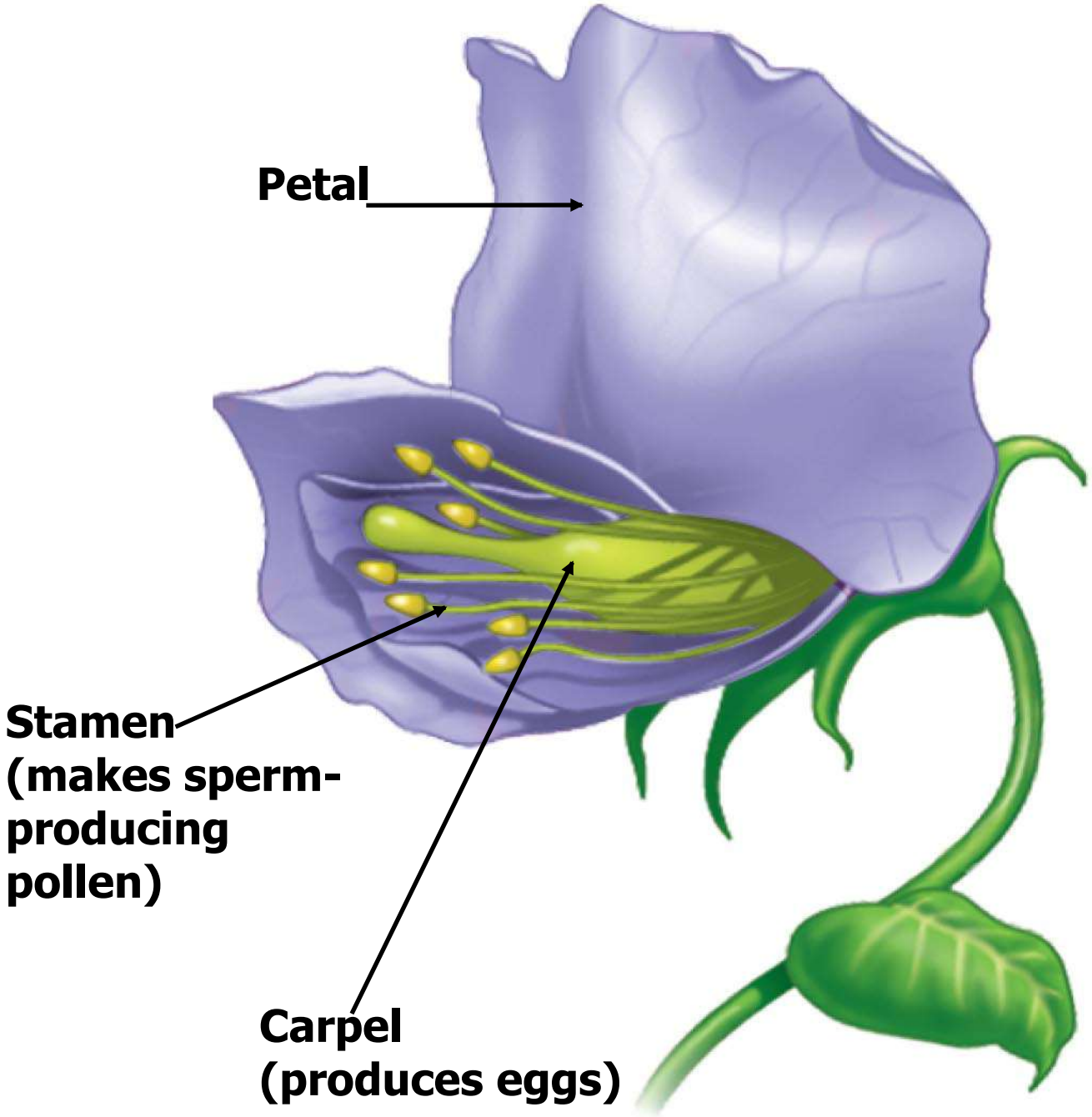

Figure 9.2

- A **character** is a heritable feature that varies among individuals.
- A **trait** is a variant of a character.
- Each of the characters Mendel studied occurred in two distinct forms.

- Mendel
  - Created true-breeding varieties of plants
  - Crossed two different true-breeding varieties
- **Hybrids** are the offspring of two different true-breeding varieties.
  - The parental plants are the **P generation**.
  - Their hybrid offspring are the **F_1 generation**.
  - A cross of the F_1 plants forms the **F_2 generation**.

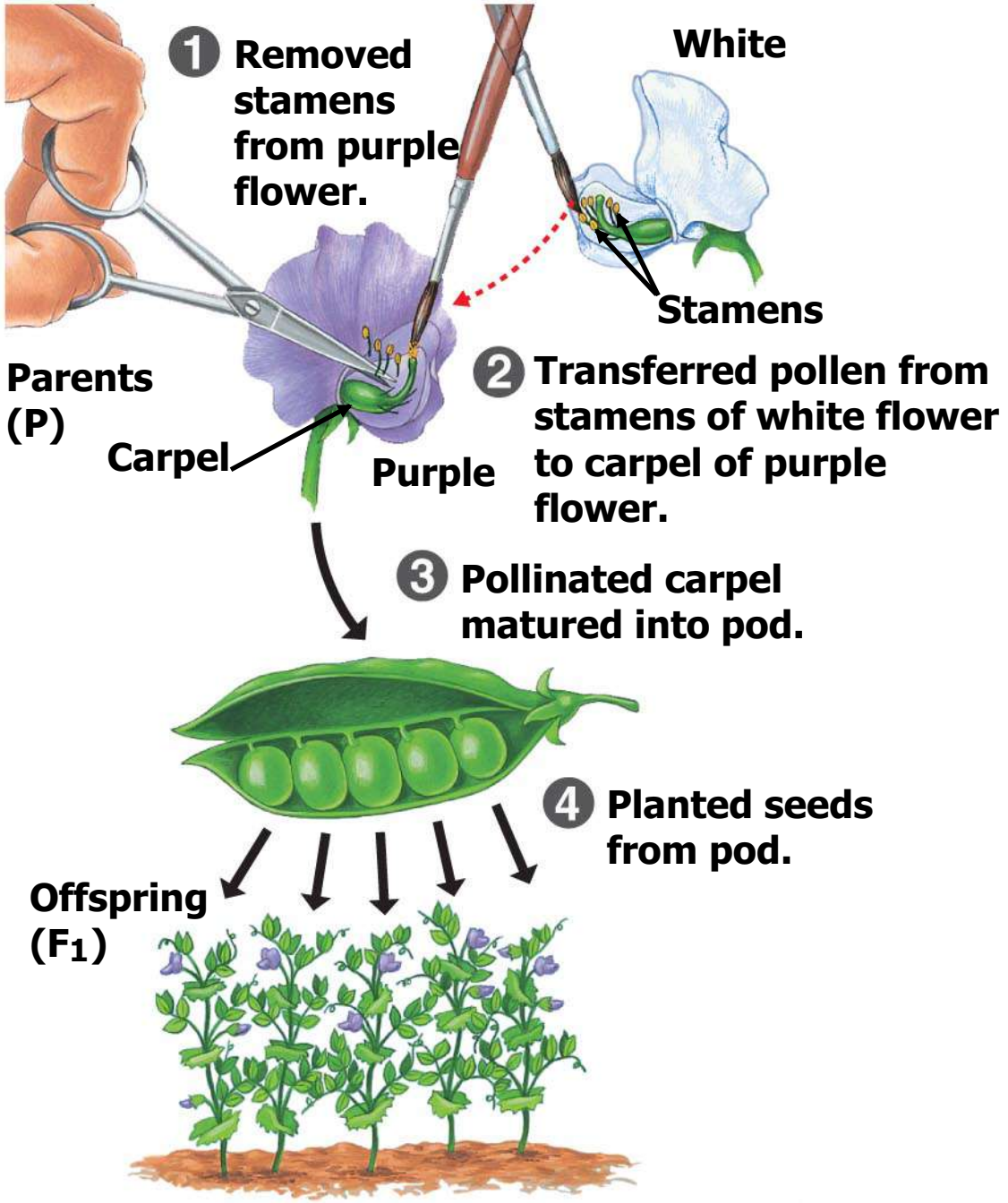

Figure 9.3-3

Mendel's Law of Segregation

- Mendel performed many experiments.
- He tracked the inheritance of characters that occur as two alternative traits.

	Dominant	Recessive
Flower color	Purple	White
Flower position	Axial	Terminal
Seed color	Yellow	Green
Seed shape	Round	Wrinkled

	Dominant	Recessive
Pod shape	Inflated	Constricted
Pod color	Green	Yellow
Stem length	Tall	Dwarf

Figure 9.4

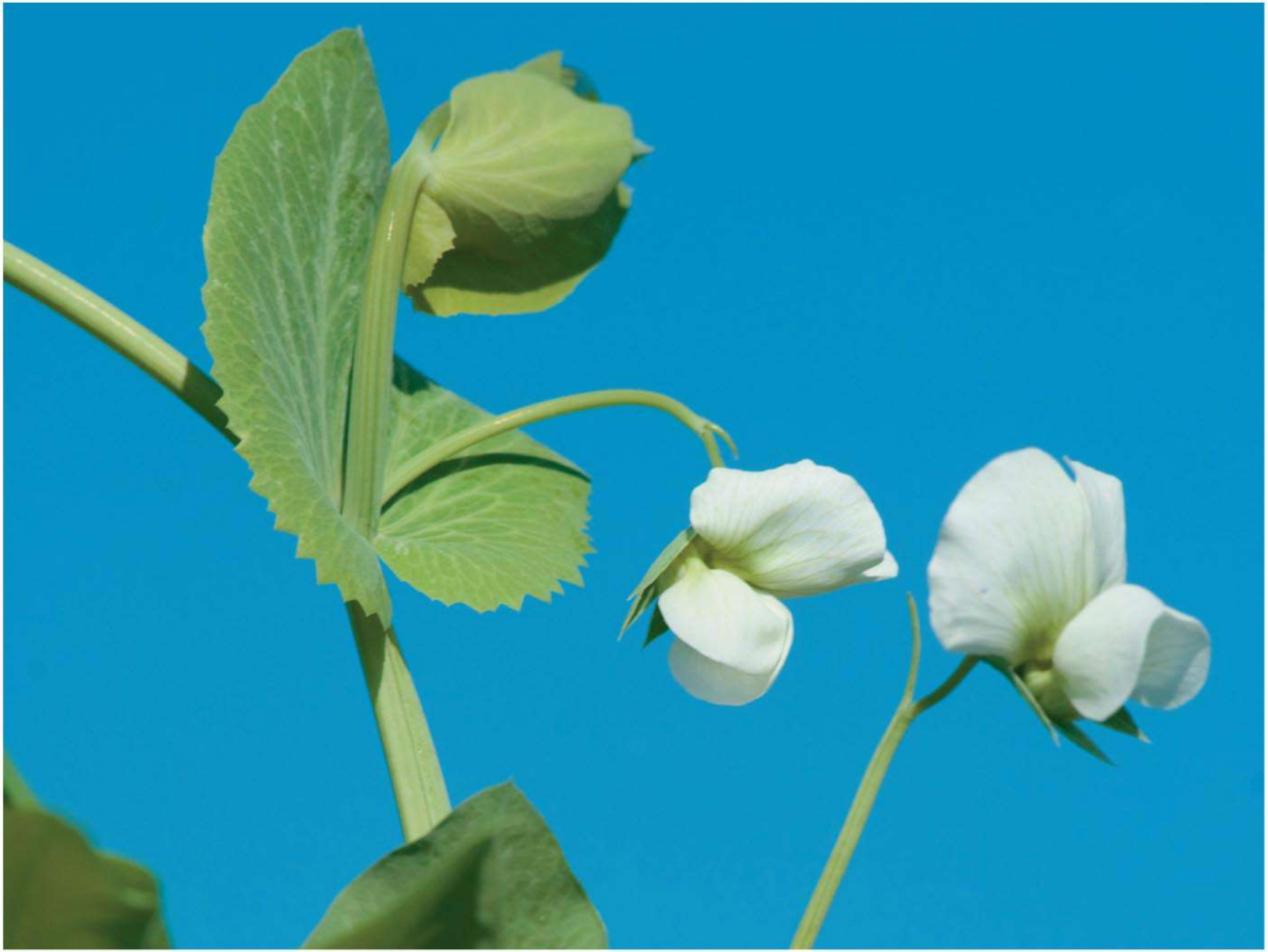

Figure 9.4

Monohybrid Crosses

- A monohybrid cross is a cross between parent plants that differ in only one character.

PLAY **Blast Animation: Single-Trait Crosses**

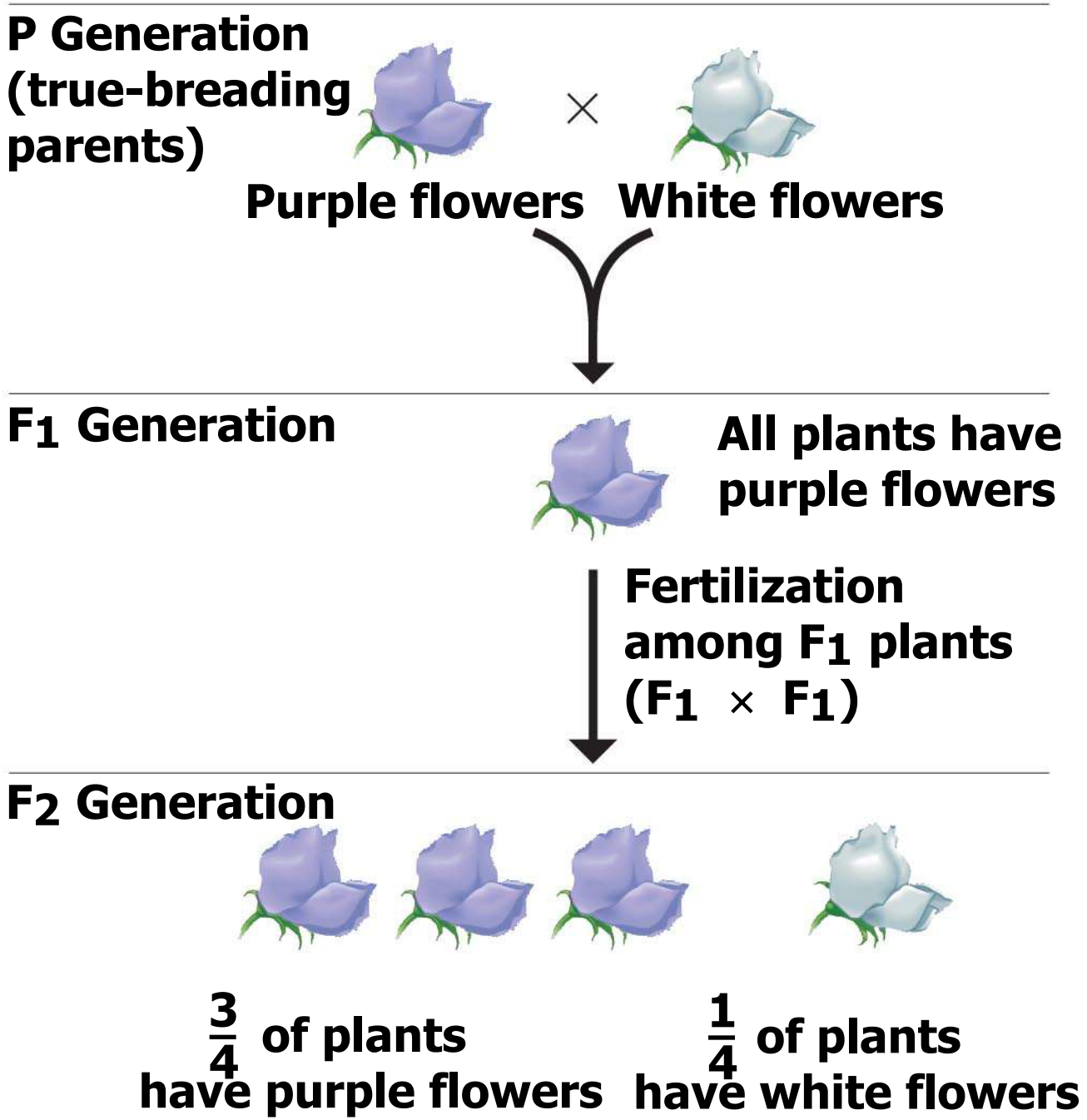

Figure 9.5-3

- Mendel developed four hypotheses from the monohybrid cross:
  1. There are alternative versions of genes, called **alleles**.
  2. For each character, an organism inherits two alleles, one from each parent.
     - An organism is **homozygous** for that gene if both alleles are identical.
     - An organism is **heterozygous** for that gene if the alleles are different.

3. If two alleles of an inherited pair differ

- The allele that determines the organism's appearance is the **dominant allele**
- The other allele, which has no noticeable effect on the appearance, is the **recessive allele**

4. Gametes carry only one allele for each inherited character.

- The two members of an allele pair segregate (separate) from each other during the production of gametes.
- This statement is the **law of segregation**.

- Do Mendel's hypotheses account for the 3:1 ratio he observed in the F_2 generation?
- A **Punnett square** highlights the four possible combinations of gametes and offspring that result from each cross.

PLAY **Blast Animation: Genetic Variation: Fusion of Gametes**

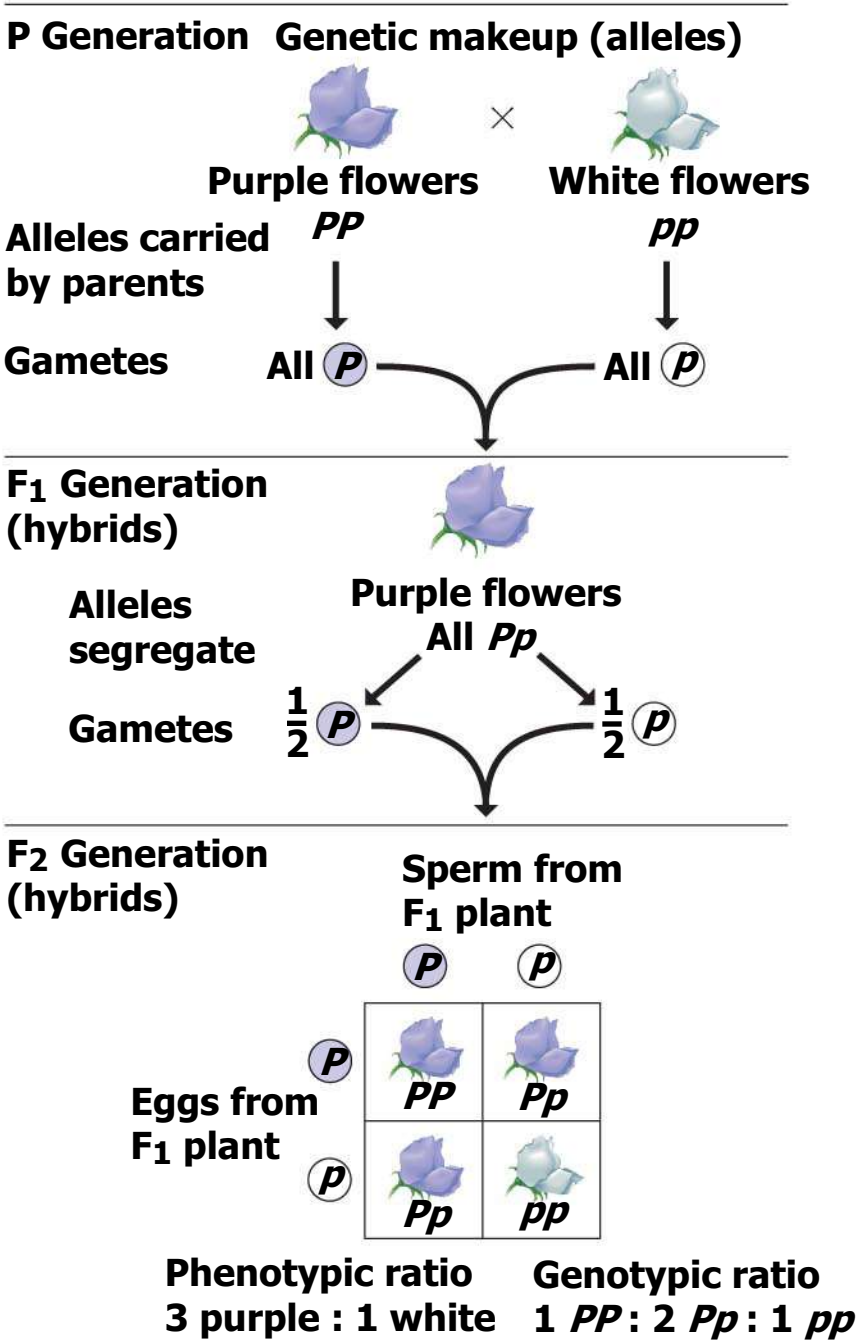

Figure 9.6-3

- Geneticists distinguish between an organism's physical traits and its genetic makeup.
  - An organism's physical traits are its **phenotype**.
  - An organism's genetic makeup is its **genotype**.

Genetic Alleles and Homologous Chromosomes

- Homologous chromosomes have
  - Genes at specific loci
  - Alleles of a gene at the same locus

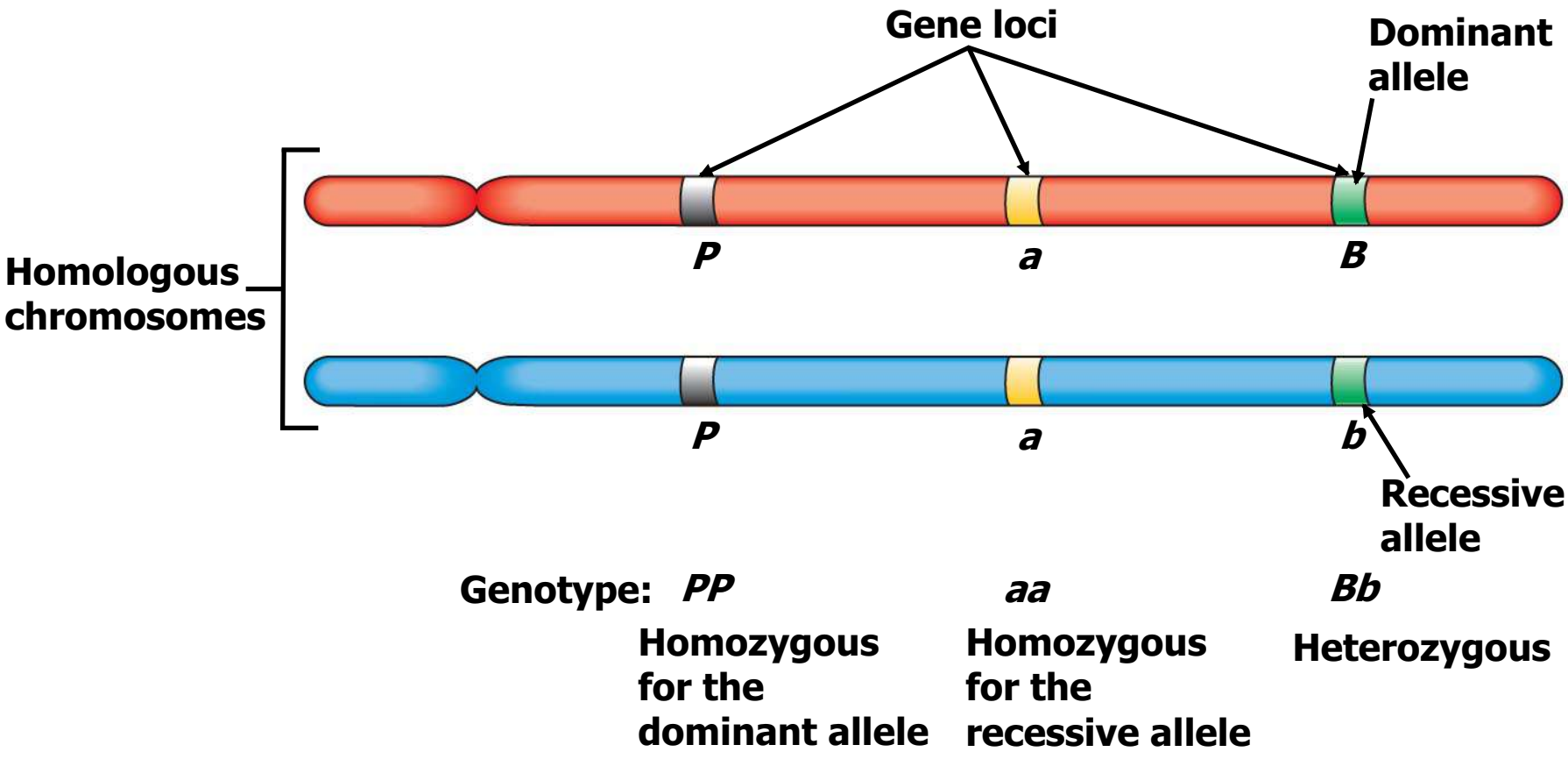

Figure 9.7

Mendel's Law of Independent Assortment

- A **dihybrid cross** is the crossing of parental varieties differing in two characters.
- What would result from a dihybrid cross? Two hypotheses are possible:
  1. Dependent assortment
  2. Independent assortment

PLAY **Blast Animation: Two-Trait Crosses**

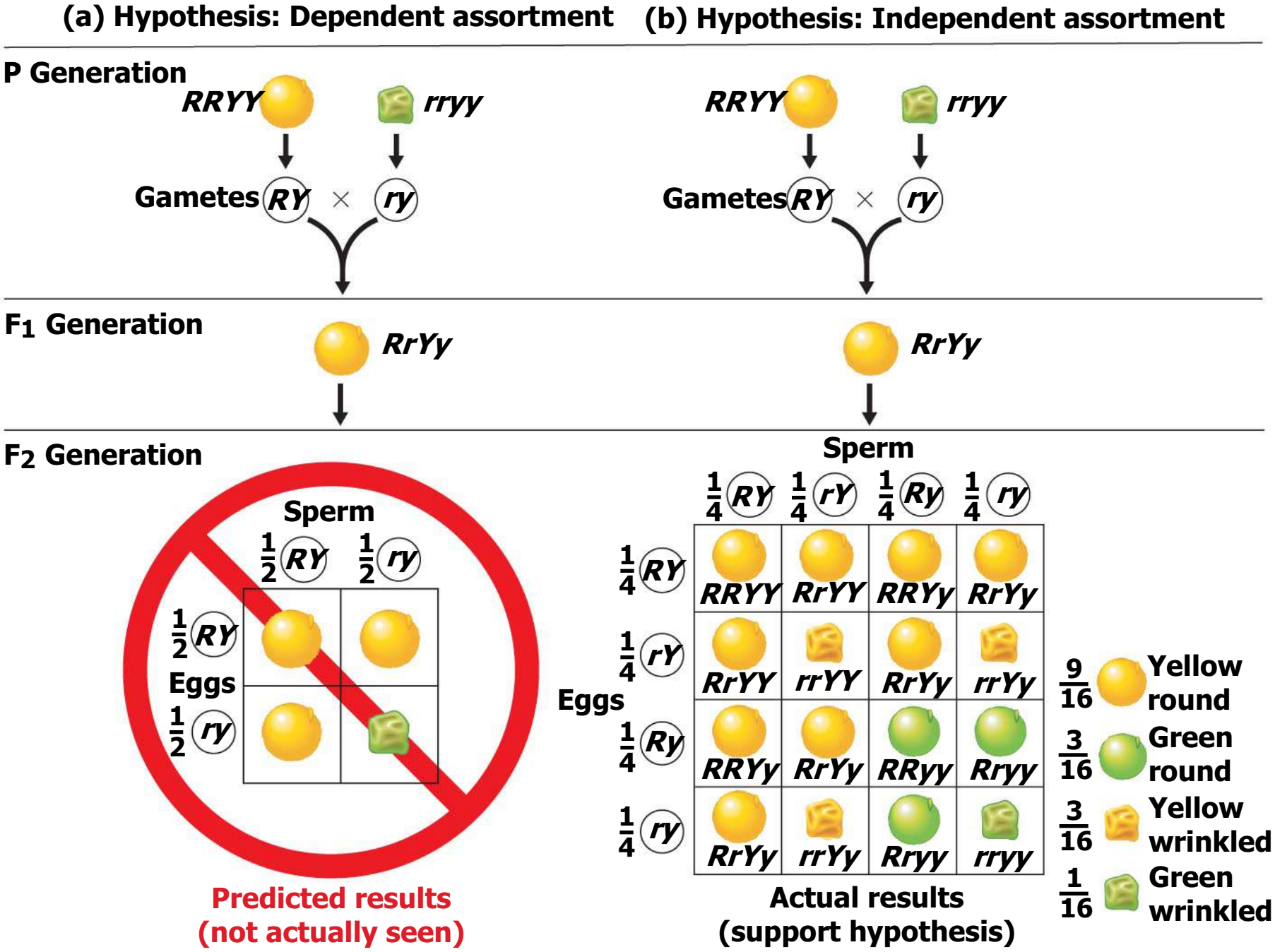
(a) Hypothesis: Dependent assortment
(b) Hypothesis: Independent assortment
P Generation
RRYY
rryy
Gametes
RY
ry
F1 Generation
RrYy
F2 Generation
Sperm
1/2 RY
1/2 ry
Eggs
Predicted results
(not actually seen)
1/4 RY
1/4 rY
1/4 Ry
1/4 ry
RRYY
RrYY
RRYy
RrYy
rrYY
rrYy
RRyy
Rryy
rryy
Actual results
(support hypothesis)
9/16 Yellow round
3/16 Green round
3/16 Yellow wrinkled
1/16 Green wrinkled
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Figure 9.8

Figure 9.8

- Mendel's dihybrid cross supported the hypothesis that each pair of alleles segregates independently of the other pairs during gamete formation.
- Thus, the inheritance of one character has no effect on the inheritance of another.
- This is the **law of independent assortment**.
- Independent assortment is also seen in two hereditary characters in Labrador retrievers.

Blind dog

Blind dog

Phenotypes	Black coat, normal vision	Black coat, blind (PRA)	Chocolate coat, normal vision	Chocolate coat, blind (PRA)
Genotypes	*B_N_*	*B_nn*	*bbN_*	*bbnn*

(a) Possible phenotypes of Labrador retrievers

Mating of double heterozygotes (black coat, normal vision)

BbNn × *BbNn*

Phenotypic ratio of offspring	9 black coat, normal vision	3 black coat, blind (PRA)	3 chocolate coat, normal vision	1 chocolate coat, blind (PRA)

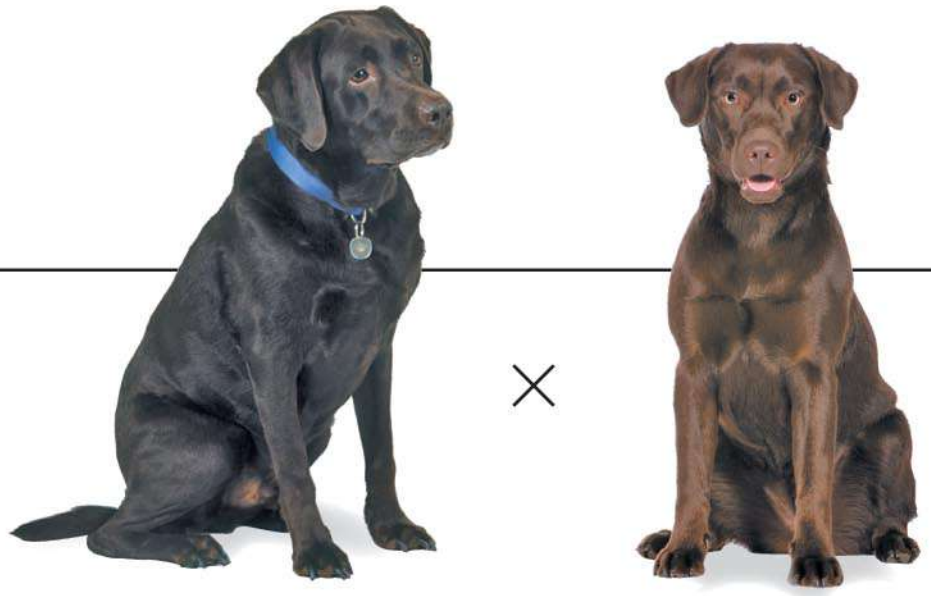
(b) A Labrador dihybrid cross

Figure 9.9

Using a Testcross to Determine an Unknown Genotype

- A **testcross** is a mating between
  - An individual of dominant phenotype (but unknown genotype)
  - A homozygous recessive individual

Testcross



Genotypes ***B_*** ***bb***

Two possible genotypes for the black dog:

BB **or** ***Bb***

Gametes

	B	***b***
b	***Bb***	***bb***

Offspring **All black** **1 black : 1 chocolate**

Figure 9.10

The Rules of Probability

- Mendel's strong background in mathematics helped him understand patterns of inheritance.
- The rule of multiplication states that the probability of a compound event is the product of the separate probabilities of the independent events.

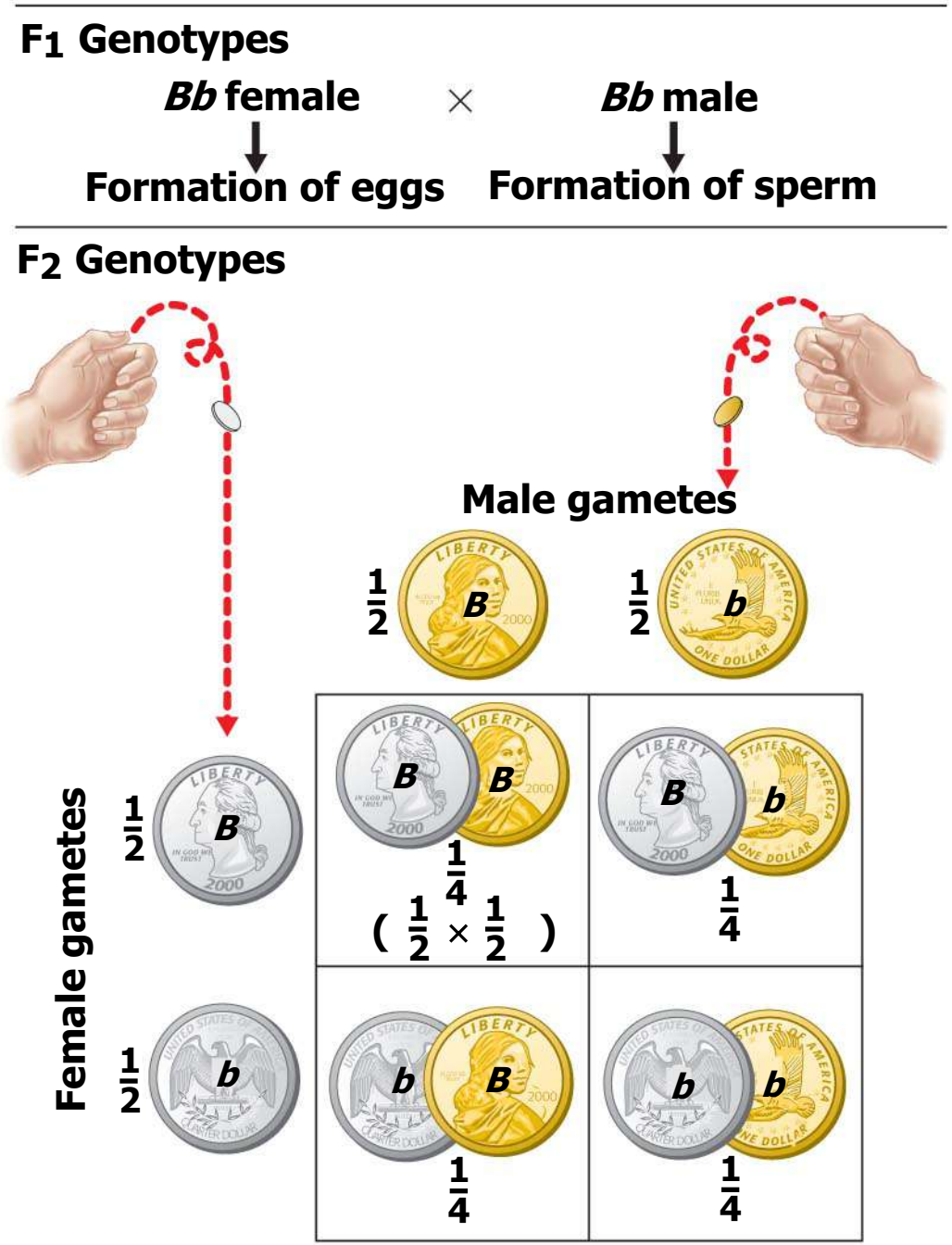


Figure 9.11

Family Pedigrees

- Mendel's principles apply to the inheritance of many human traits.

DOMINANT TRAITS

Freckles

Widow's peak

Free earlobe

RECESSIVE TRAITS

No freckles

Straight hairline

Attached earlobe

Figure 9.12

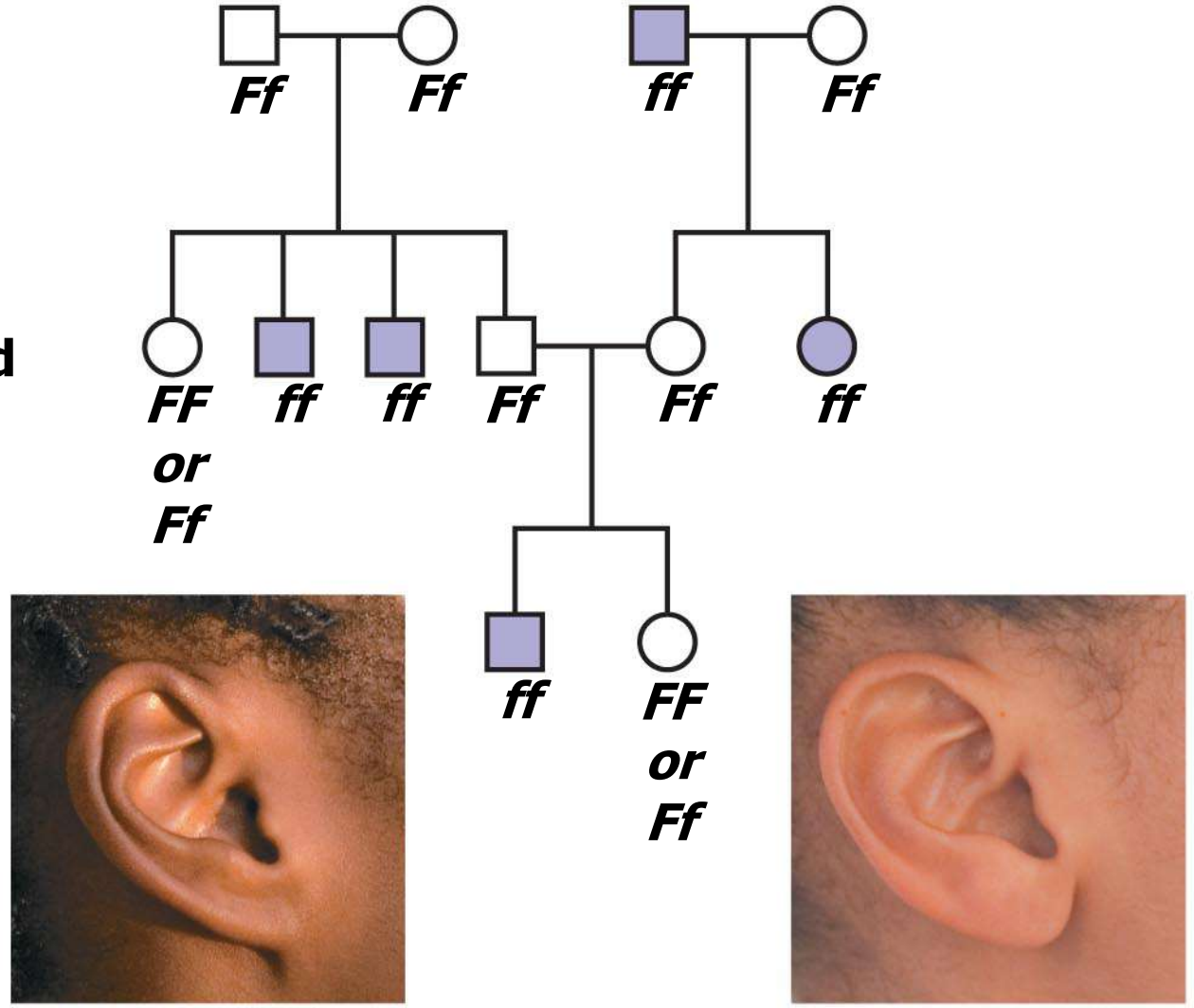
- Dominant traits are not necessarily
  - Normal or
  - More common
- **Wild-type traits** are
  - Those seen most often in nature
  - Not necessarily specified by dominant alleles

- A family pedigree
  - Shows the history of a trait in a family
  - Allows geneticists to analyze human traits

First generation (grandparents)

Second generation (parents, aunts, and uncles)

Third generation (brother and sister)



Female Male

Attached

Free

Figure 9.13

Human Disorders Controlled by a Single Gene

- Many human traits
  - Show simple inheritance patterns
  - Are controlled by single genes on autosomes

Table 9.1	Some Autosomal Disorders in Humans	
Disorder	**Major Symptoms**	**Incidence**
Recessive Disorders		
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated	$\frac{1}{1,800}$ European-Americans
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation unless treated	$\frac{1}{10,000}$ in U.S. and Europe
Sickle-cell disease	Sickled red blood cells; damage to many tissues	$\frac{1}{500}$ African-Americans
Tay Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ European Jews
Dominant Disorders		
Achondroplasia	Dwarfism	$\frac{1}{25,000}$
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$

Table 9.1

Recessive Disorders

- Most human genetic disorders are recessive.
- Individuals who have the recessive allele but appear normal are **carriers** of the disorder.

Parents

Hearing *Dd* × Hearing *Dd*

Offspring

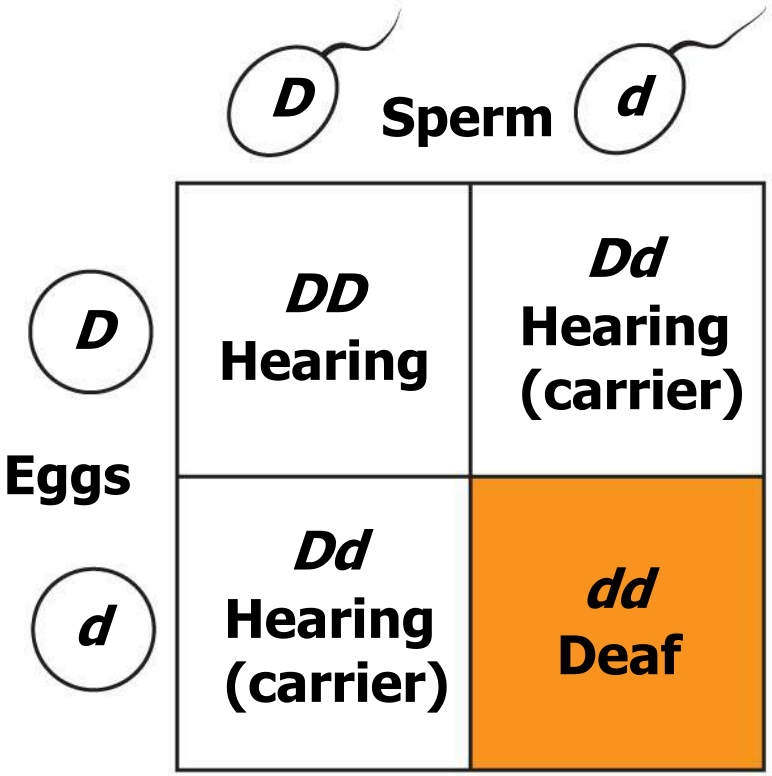


Figure 9.14

- Cystic fibrosis
  - Is the most common lethal genetic disease in the United States
  - Is caused by a recessive allele carried by about one in 25 people of European ancestry
- Prolonged geographic isolation of certain populations can lead to **inbreeding**, the mating of close relatives.
  - Inbreeding increases the chance of offspring that are homozygous for a harmful recessive trait.

Dominant Disorders

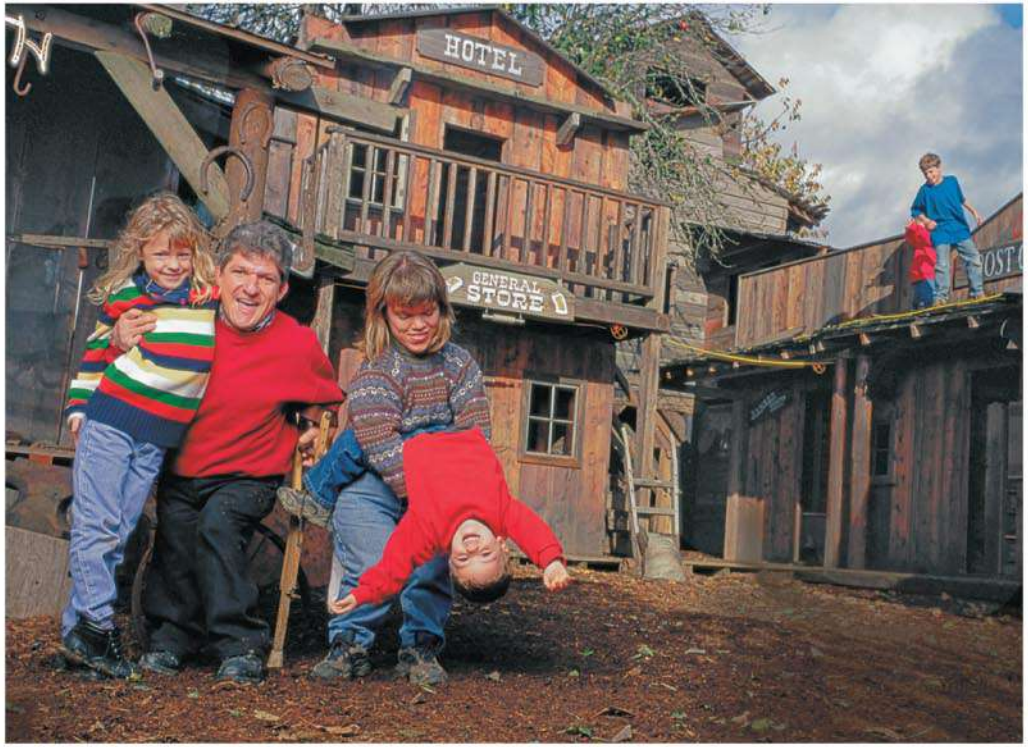
- Some human genetic disorders are dominant.
  - **Huntington's disease**, which leads to degeneration of the nervous system, does not begin until middle age.
  - **Achondroplasia** is a form of dwarfism.
    - The homozygous dominant genotype causes death of the embryo.
    - Thus, only heterozygotes have this disorder.

Parents

Normal (no achondroplasia) × Dwarf (achondroplasia)

dd × *Dd*

Eggs	*d* (Sperm)	*d* (Sperm)
D	***Dd*** **Dwarf**	***Dd*** **Dwarf**
d	***dd*** **Normal**	***dd*** **Normal**



Molly Jo Matt Amy Zachary Jake Jeremy

Figure 9.16

The Process of Science: What Is the Genetic Basis of Hairless Dogs?

- **Observation**: Dogs come in a wide variety of physical types.
- **Question**: What is the genetic basis for the hairless phenotype?
- **Hypothesis**: A comparison of genes of coated and hairless dogs would identify the gene or genes responsible.

- **Prediction**: A mutation in a single gene accounts for the hairless appearance.
- **Experiment**: Compared DNA sequences of 140 hairless dogs from 3 breeds with 87 coated dogs from 22 breeds.
- **Results**: Every hairless dog, but no coated dogs, had a single change in a single gene.

Figure 9.17

Genetic Testing

- Today many tests can detect the presence of disease-causing alleles.
- Most genetic testing is performed during pregnancy.
  - Amniocentesis collects cells from amniotic fluid.
  - Chorionic villus sampling removes cells from placental tissue.
- Genetic counseling helps patients understand the results and implications of genetic testing.

VARIATIONS ON MENDEL'S LAWS

- Some patterns of genetic inheritance are not explained by Mendel's laws.

Incomplete Dominance in Plants and People

- In **incomplete dominance**, F_1 hybrids have an appearance in between the phenotypes of the two parents.

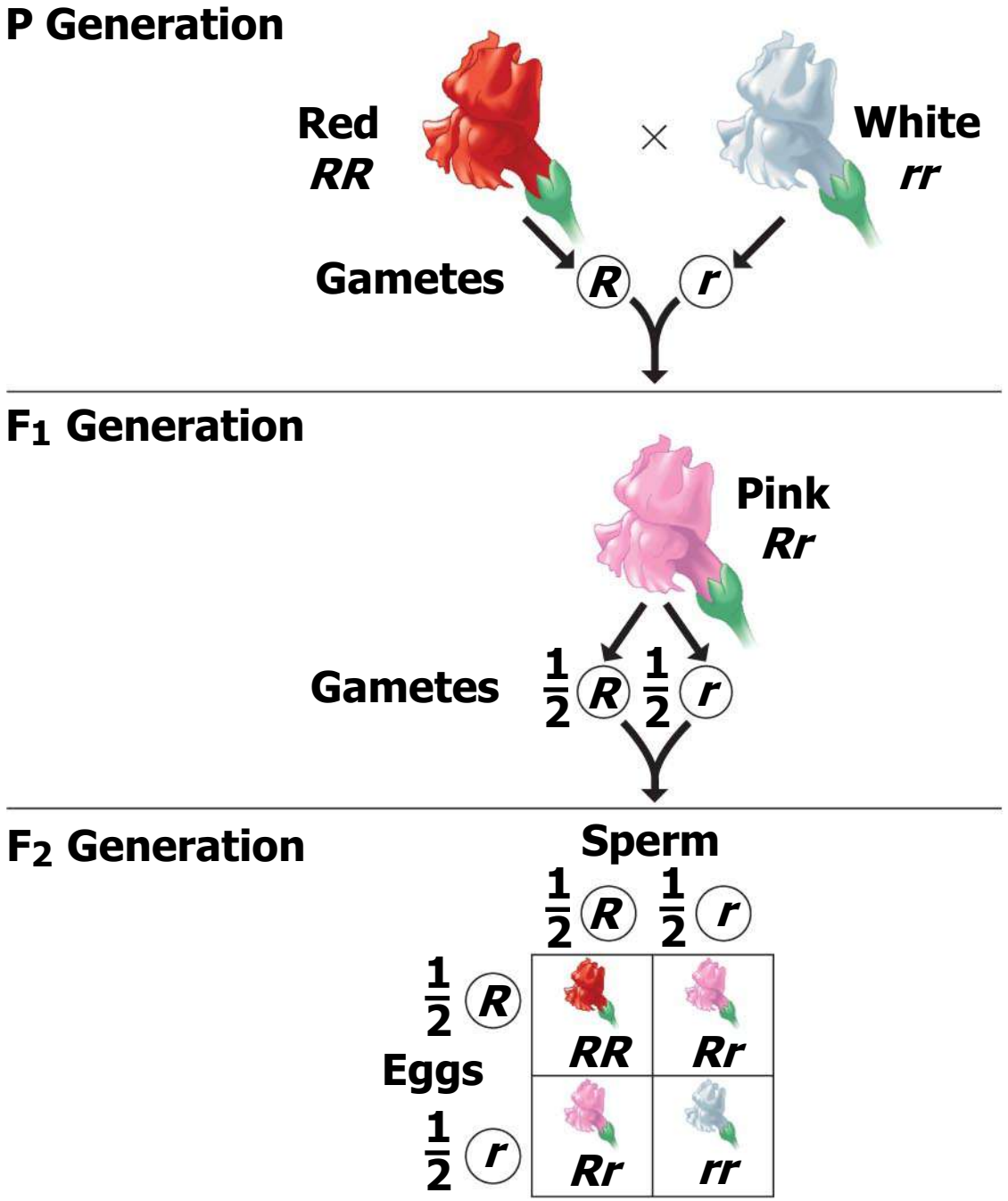


Figure 9.18-3

- **Hypercholesterolemia**
  - Is characterized by dangerously high levels of cholesterol in the blood.
  - Is a human trait that is incompletely dominant.
  - Heterozygotes have blood cholesterol levels about twice normal.
  - Homozygotes have blood cholesterol levels about five times normal.

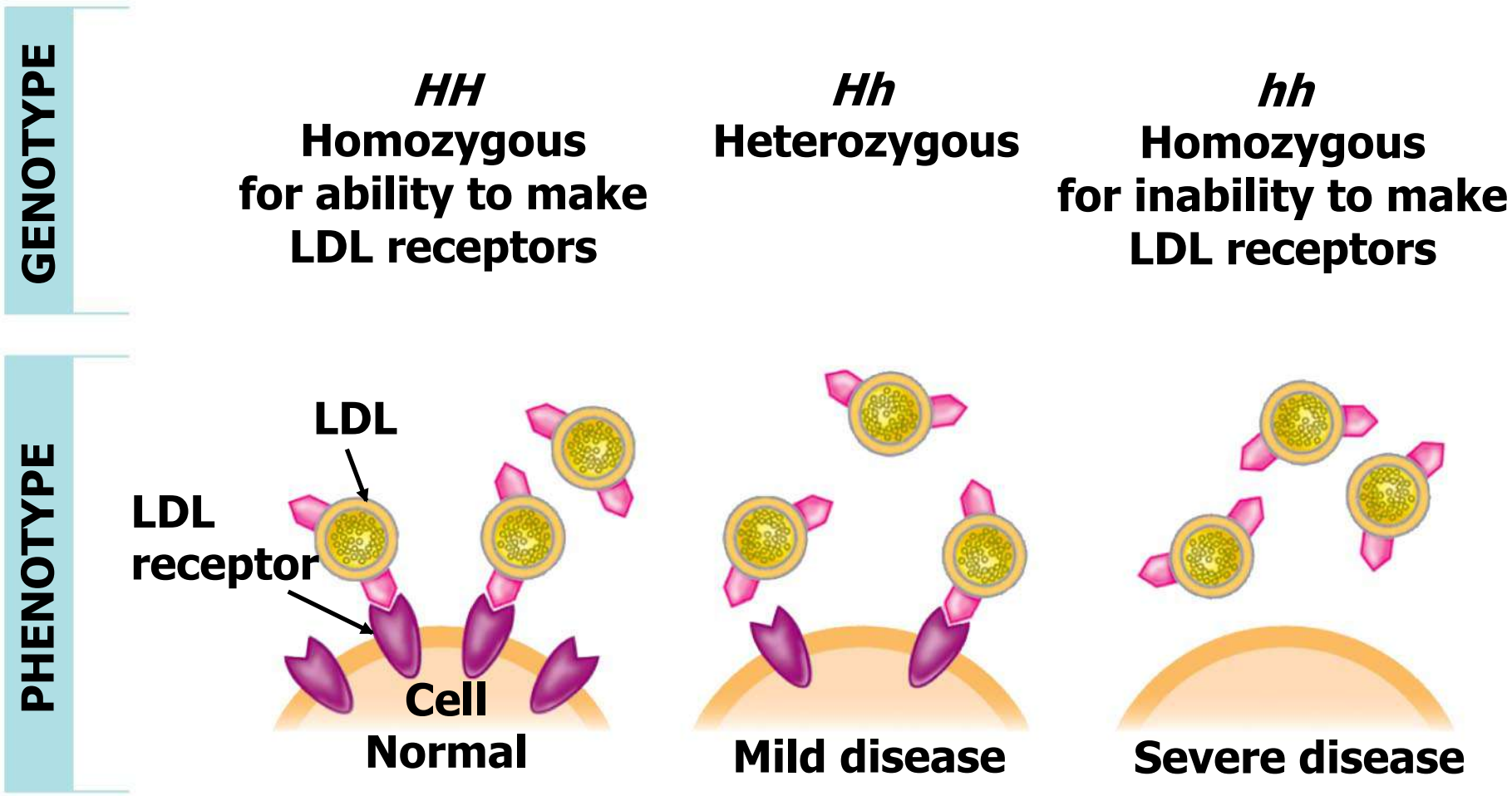


Figure 9.19

ABO Blood Groups: An Example of Multiple Alleles and Codominance

- The **ABO blood groups** in humans are an example of multiple alleles.

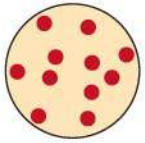
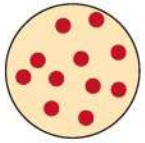
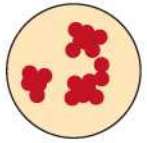
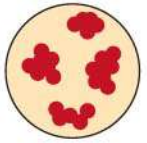
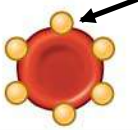
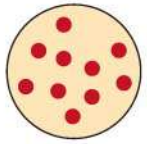
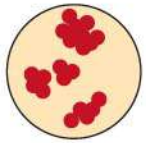
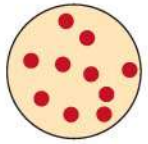
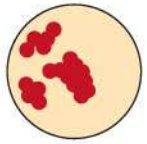
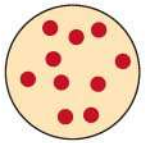
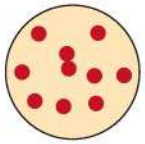
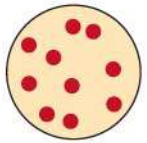
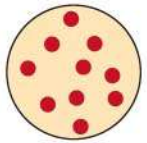
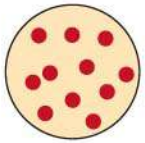
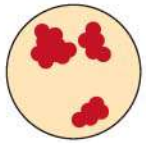
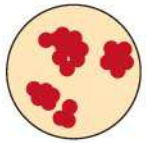
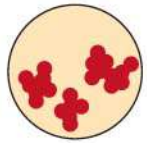
Blood Group (Phenotype)	Genotypes	Red Blood Cells	Antibodies Present in Blood	Reactions When Blood from Groups Below Is Mixed with Antibodies from Groups at Left			
				O	A	B	AB
A	I^AI^A or I^Ai	Carbohydrate A 	Anti-B				
B	I^BI^B or I^Bi	Carbohydrate B 	Anti-A				
AB	I^AI^B		—				
O	ii		Anti-A Anti-B				

Figure 9.20

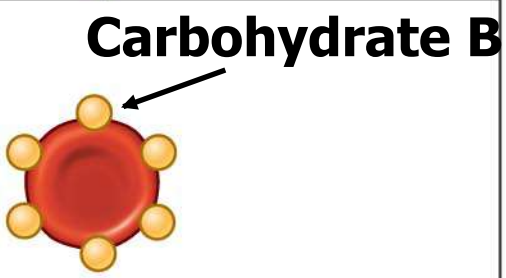
Blood Group (Phenotype)	Genotypes	Red Blood Cells
A	I^AI^A or I^Ai	
B	I^BI^B or I^Bi	
AB	I^AI^B	
O	ii	

Figure 9.20a

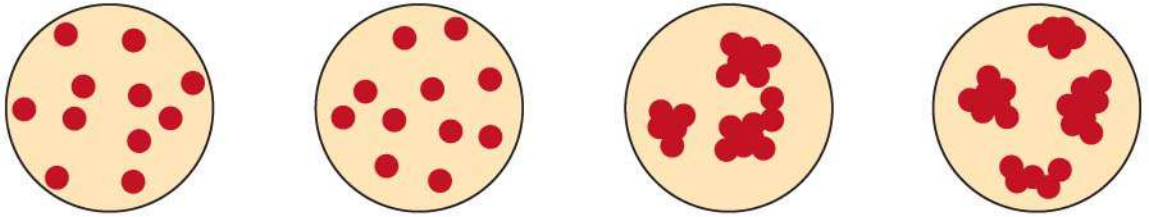
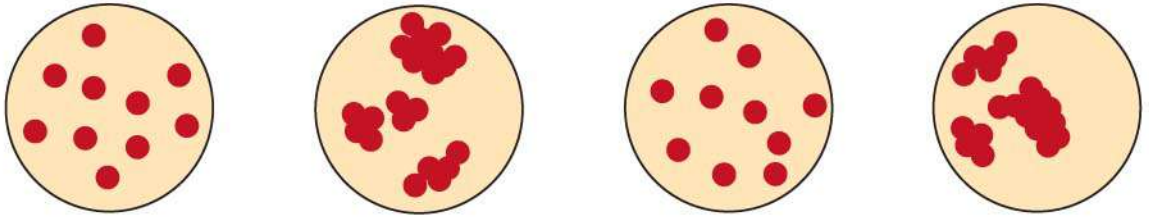
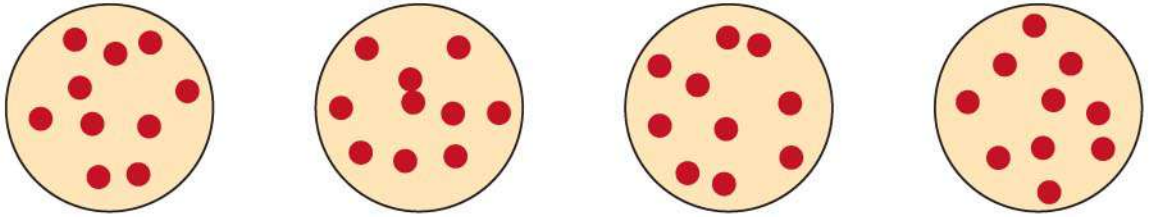
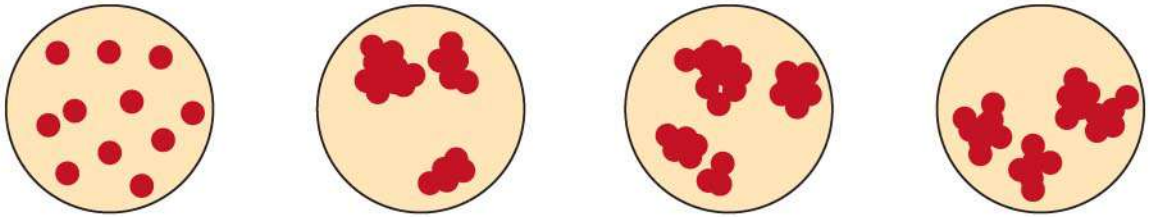
Antibodies Present in Blood	Reactions When Blood from Groups Below Is Mixed with Antibodies from Groups at Left			
	O	A	B	AB
Anti-B				
Anti-A				
—				
Anti-A Anti-B				

Figure 9.20b

Figure 9.20

- The immune system produces blood proteins called antibodies that can bind specifically to blood cell carbohydrates.
- Blood cells may clump together if blood cells of a different type enter the body.
- The clumping reaction is the basis of a blood-typing lab test.

Individual homozygous, for sickle-cell allele

Sickle-cell (abnormal) hemoglobin

Abnormal hemoglobin crystallizes into long flexible chains, causing red blood cells to become sickle-shaped.

Colorized SEM

Sickled cells can lead to a cascade of symptoms, such as weakness, pain, organ damage, and paralysis.

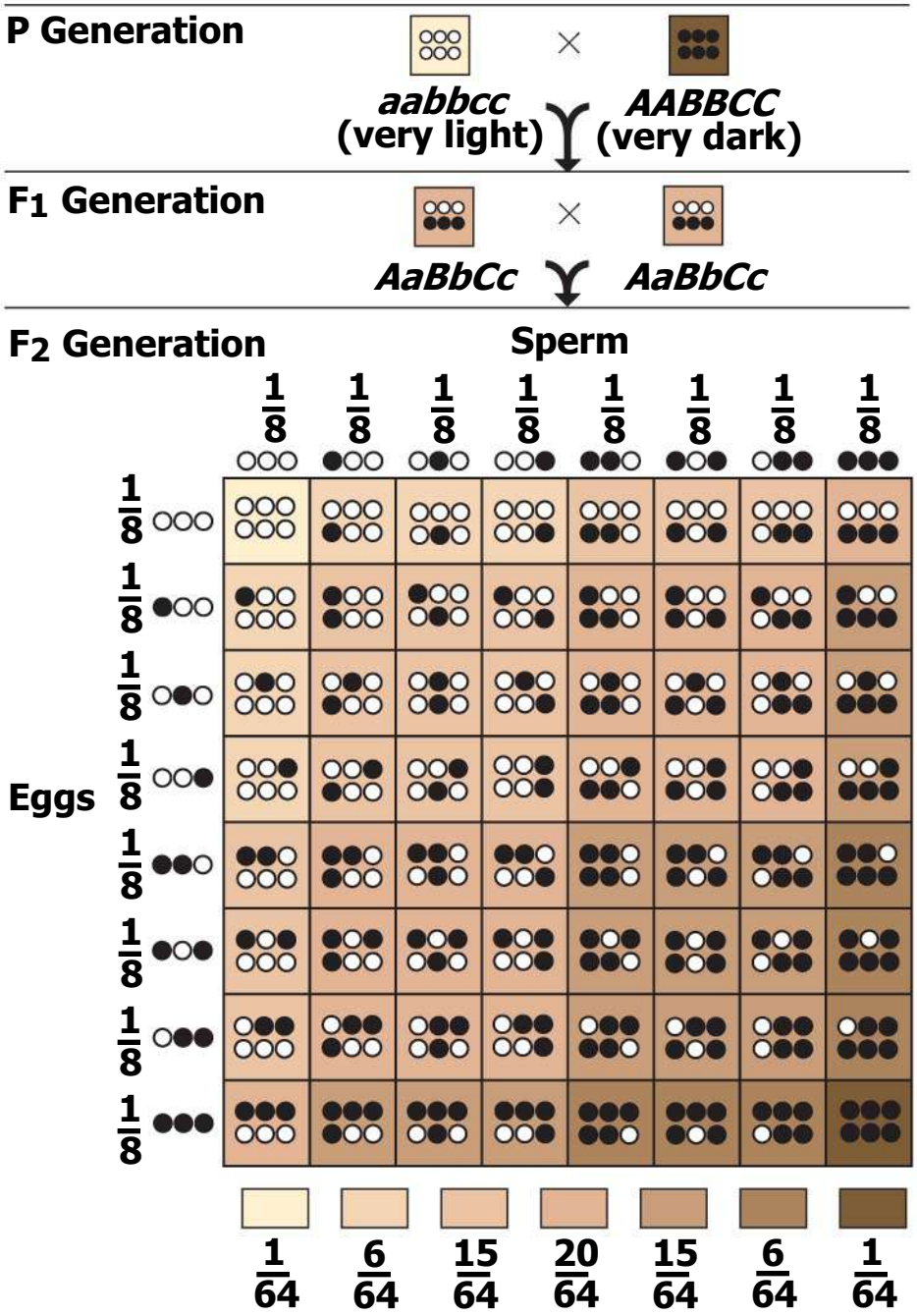
Figure 9.21

Figure 9.21

- The human blood type alleles I^A and I^B exhibit **codominance**: Both alleles are expressed in the phenotype.

Pleiotropy and Sickle-Cell Disease

- **Pleiotropy** is the impact of a single gene on more than one character.
- **Sickle-cell disease**
  - Exhibits pleiotropy
  - Results in abnormal hemoglobin production
  - Causes disk-shaped red blood cells to deform into a sickle shape with jagged edges



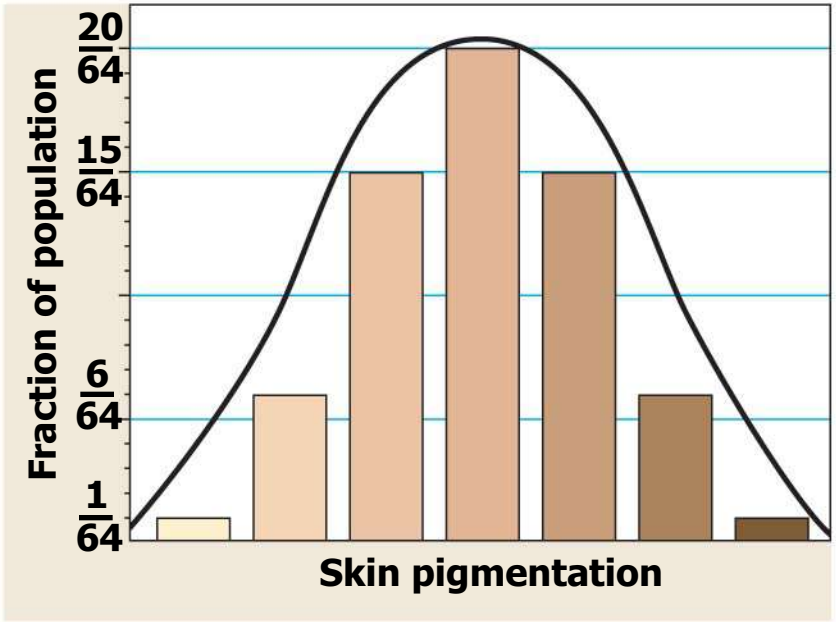


Figure 9.22

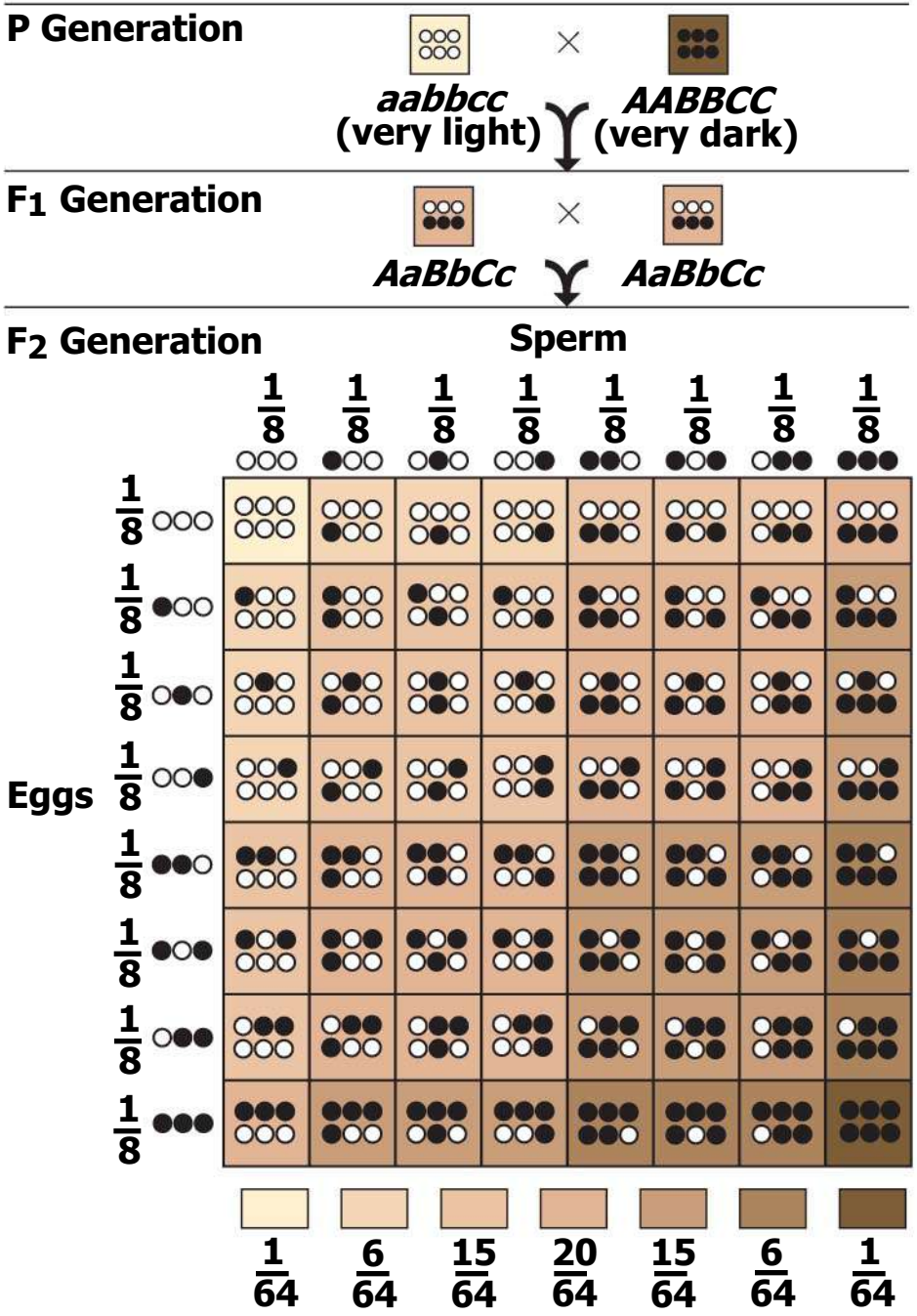


Figure 9.22a

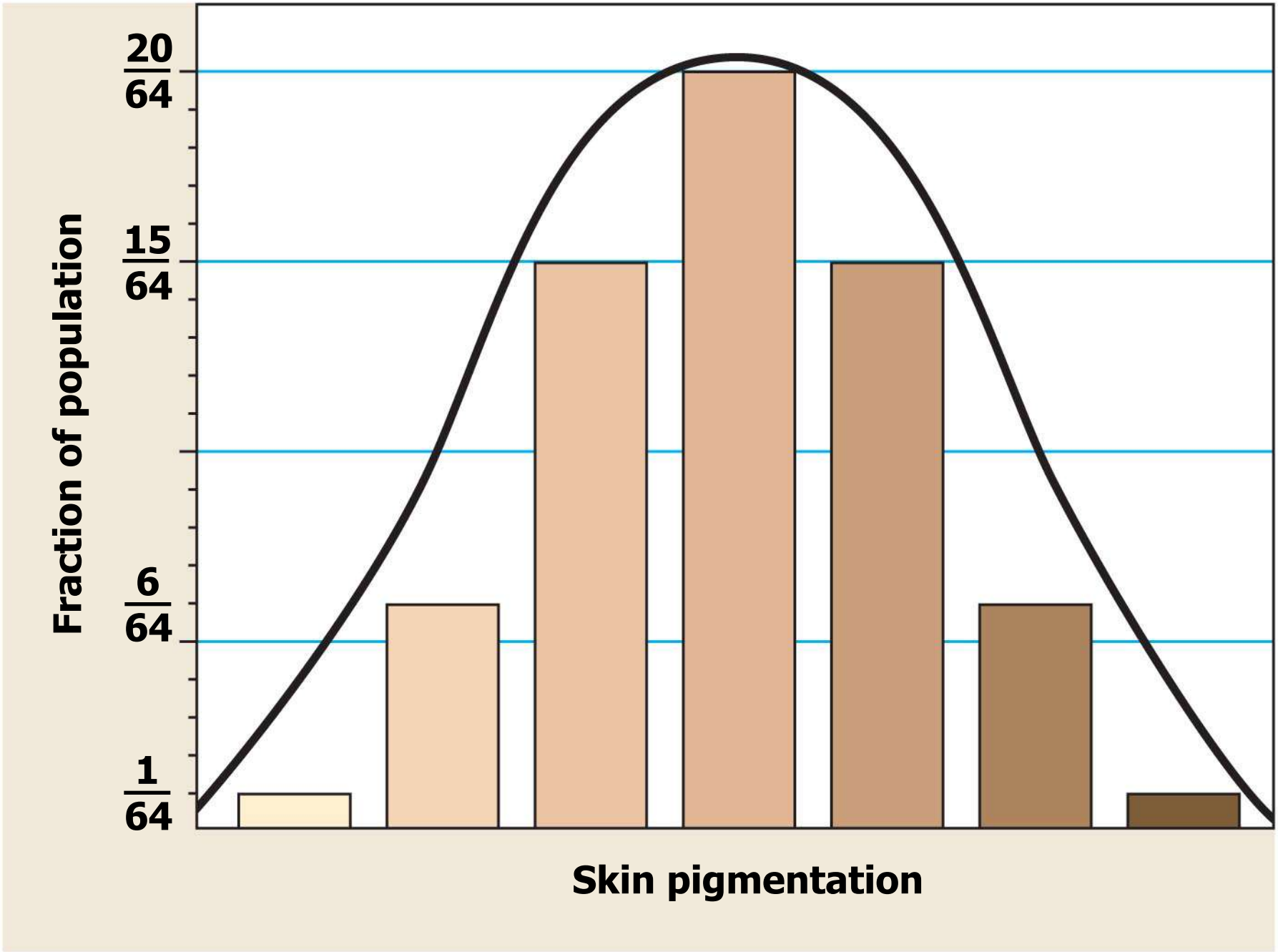


Figure 9.22b

Polygenic Inheritance

- **Polygenic inheritance** is the additive effects of two or more genes on a single phenotype.

Figure 9.23

The Role of Environment

- Many human characters result from a combination of heredity and environment.
- Only genetic influences are inherited.

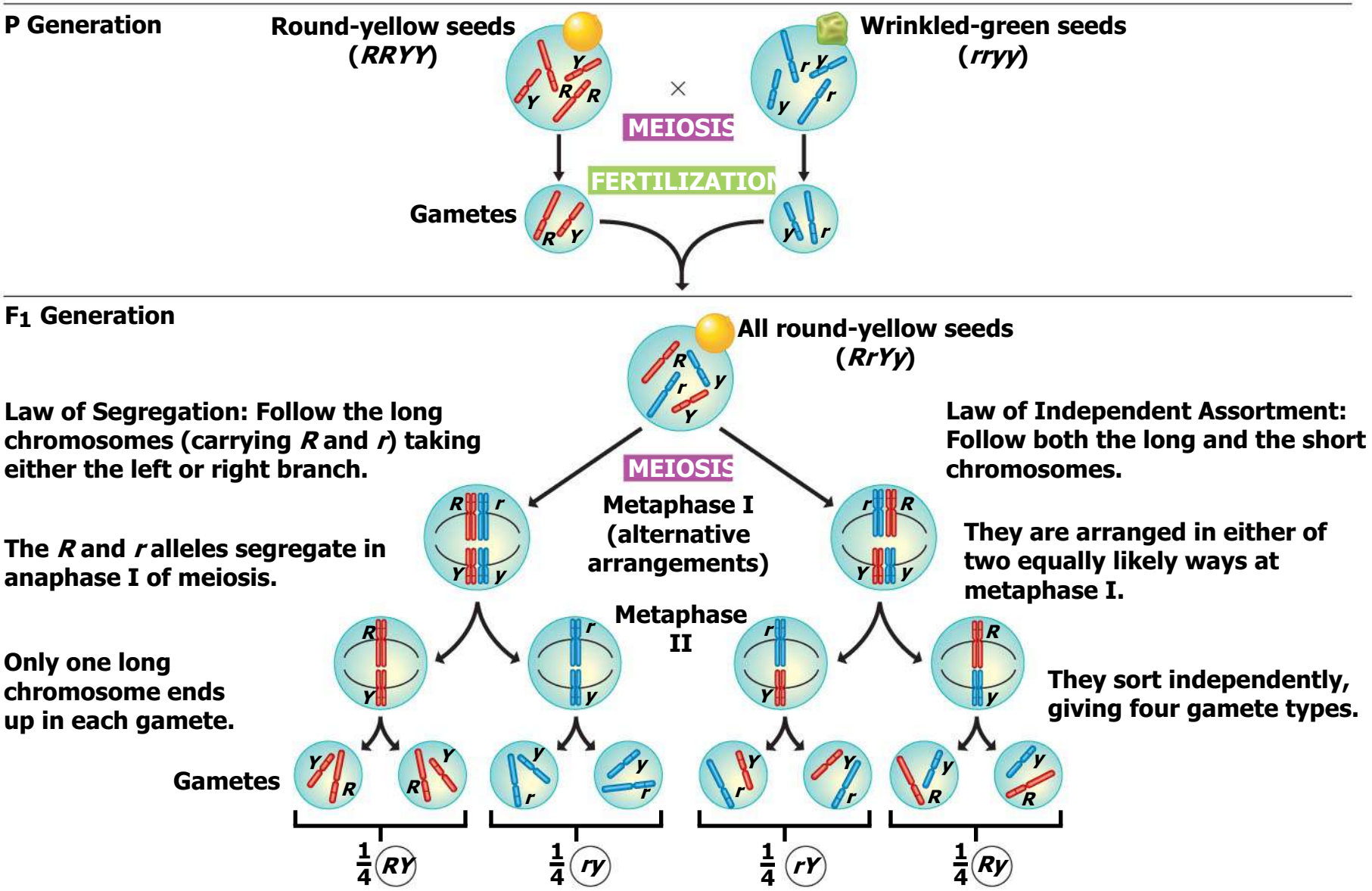


Figure 9.24-3

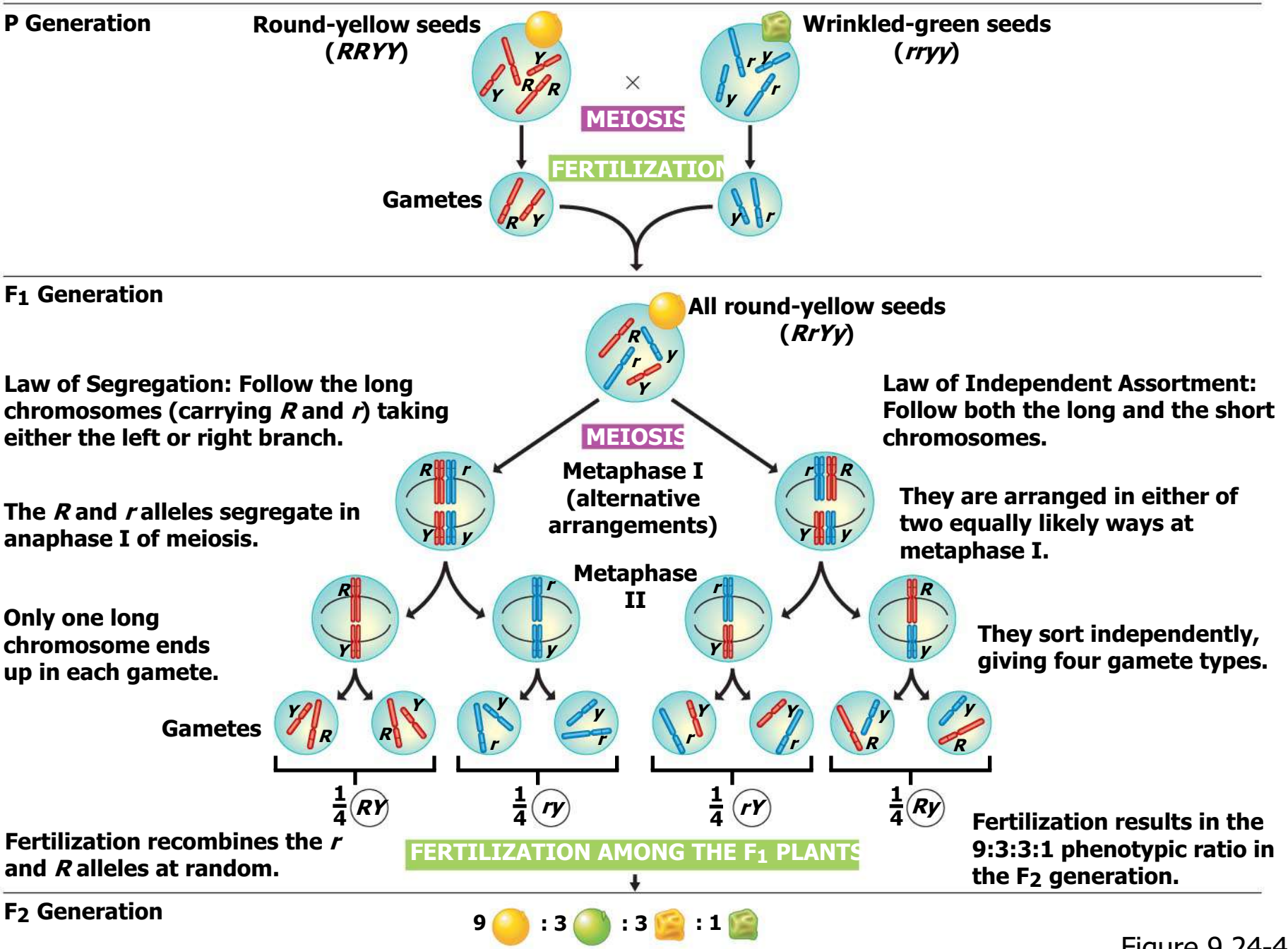


Figure 9.24-4

THE CHROMOSOMAL BASIS OF INHERITANCE

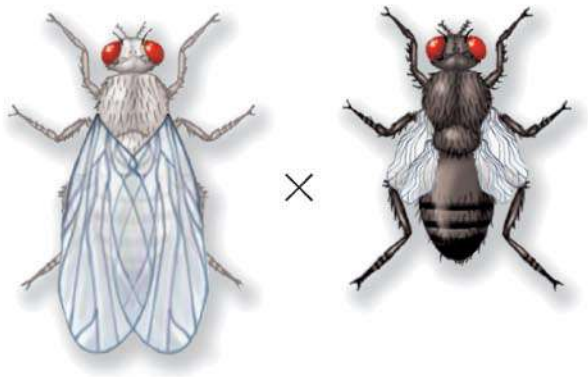
- The **chromosome theory of inheritance** states that
  - Genes are located at specific positions on chromosomes
  - The behavior of chromosomes during meiosis and fertilization accounts for inheritance patterns
- It is *chromosomes* that undergo segregation and independent assortment during meiosis and thus account for Mendel's laws.

Dihybrid testcross

Gray body, long wings (wild-type)

GgLl

Female



Black body, short wings (mutant)

ggll

Male

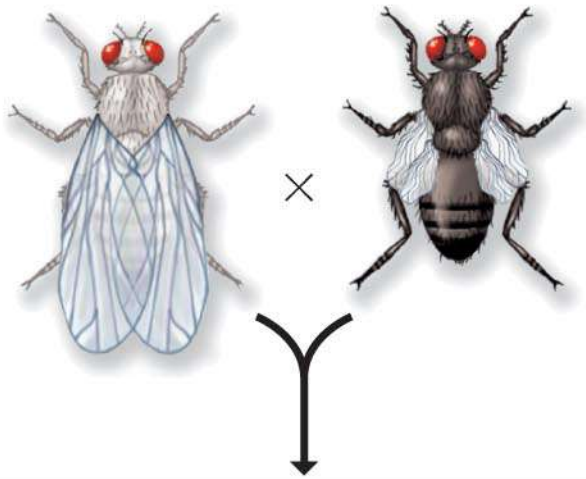
Figure 9.25-1

Dihybrid testcross

Gray body, long wings (wild-type)

GgLl

Female



Black body, short wings (mutant)

ggll

Male

Results

Offspring

Gray-long *GgLl*	Black-short *ggll*	Gray-short *Ggll*	Black-long *ggLl*
965	944	206	185
Parental phenotypes 83%		Recombinant phenotypes 17%	

Figure 9.25-2

Linked Genes

- **Linked genes**
  - Are located close together on a chromosome
  - May be inherited together
- Using the fruit fly *Drosophila melanogaster*, Thomas Hunt Morgan determined that some genes were linked based on the inheritance patterns of their traits.

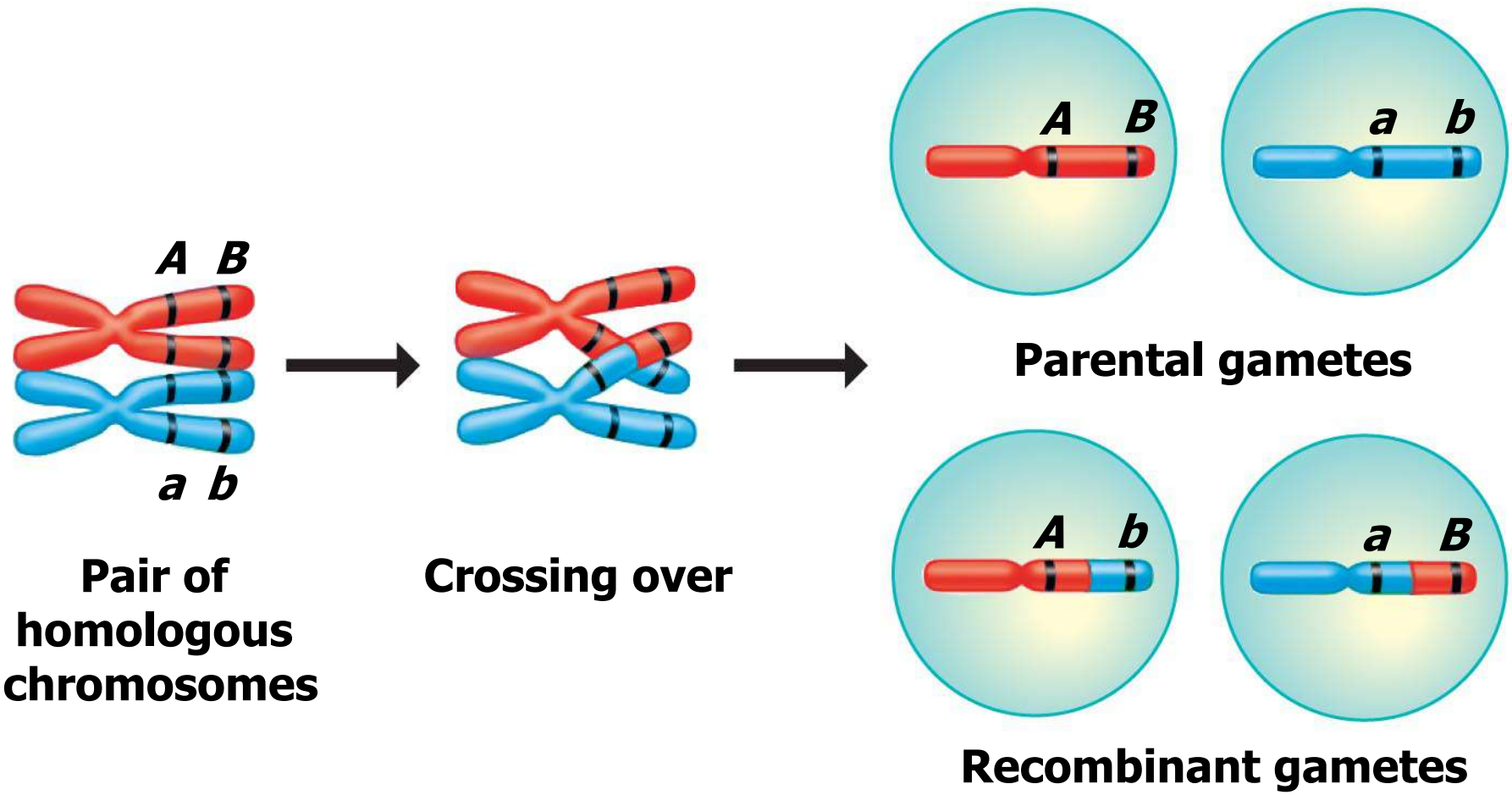


Figure 9.26

Genetic Recombination: Crossing Over

- Crossing over can
  - Separate linked alleles
  - Produce gametes with recombinant chromosomes
  - Produce offspring with recombinant phenotypes

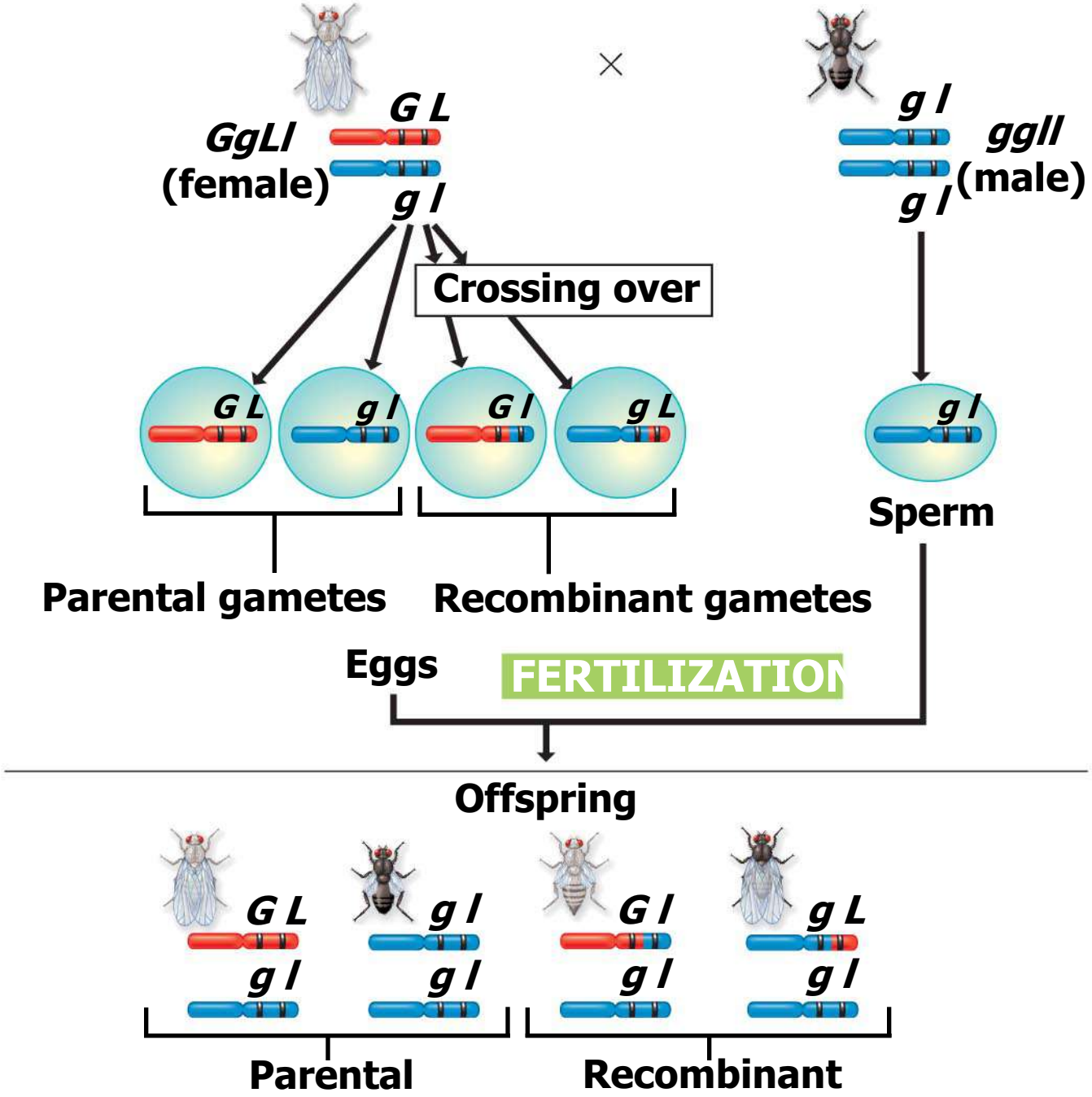


Figure 9.27

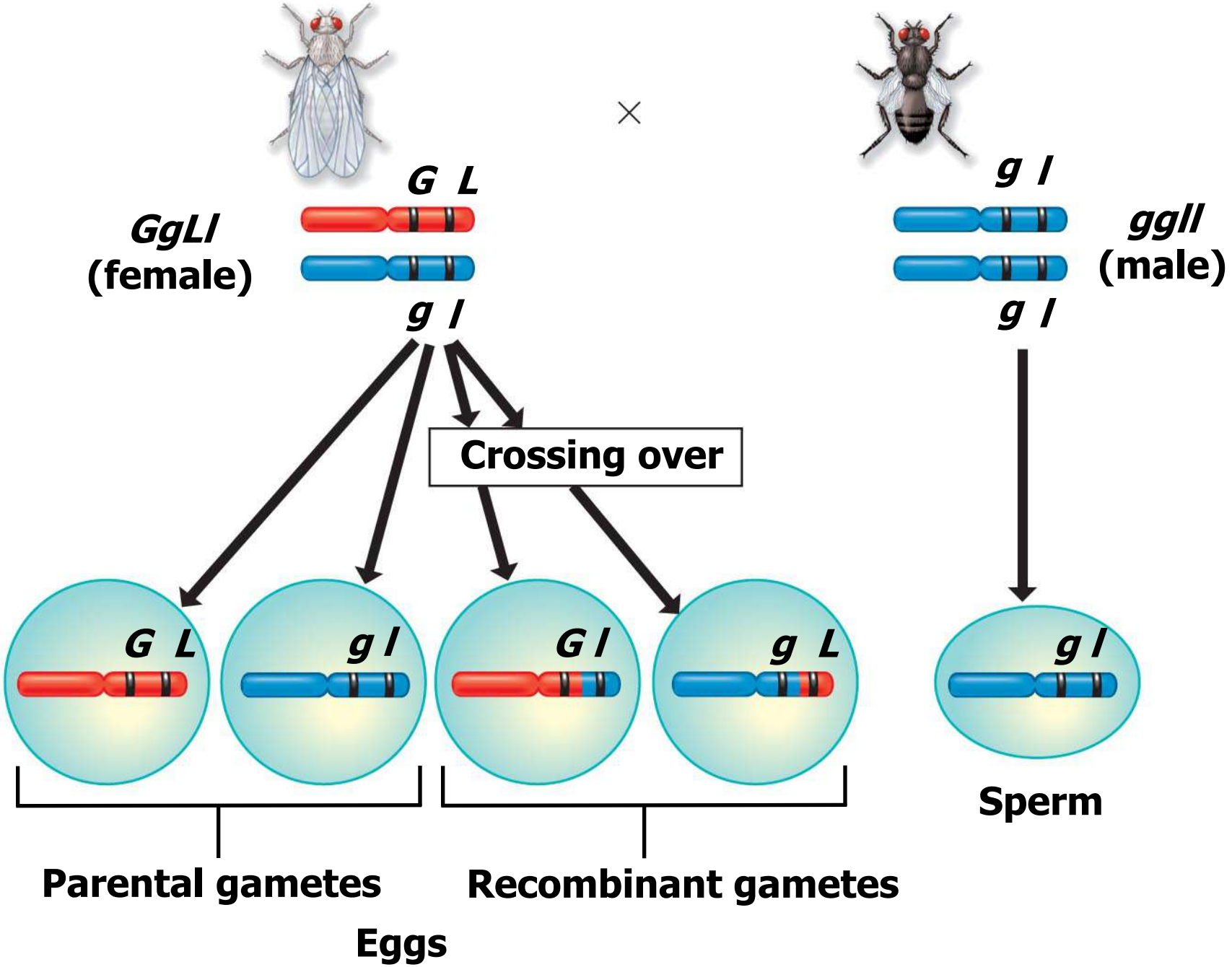


Figure 9.27a

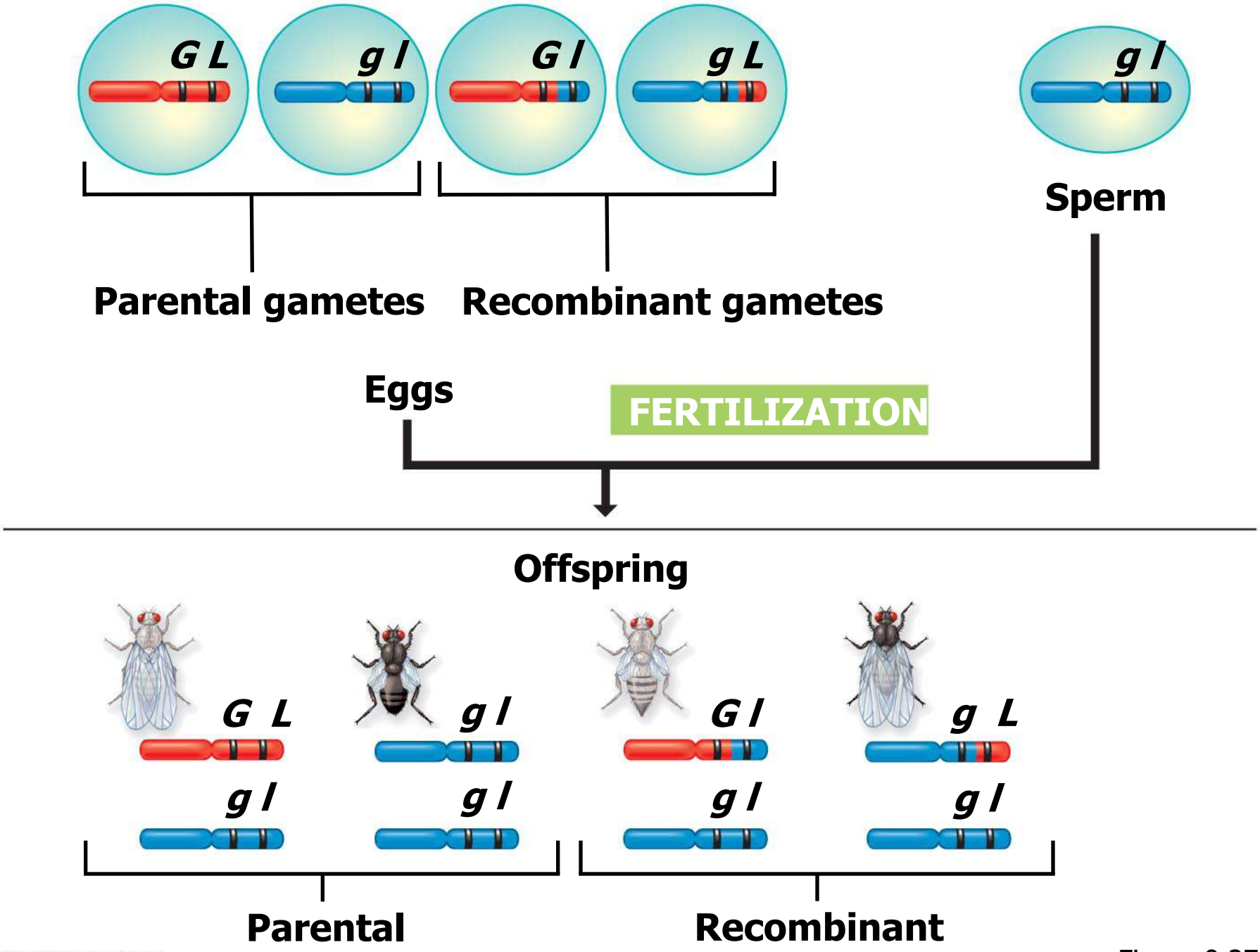


Figure 9.27b

- The percentage of recombinant offspring among the total is called the **recombination frequency**.

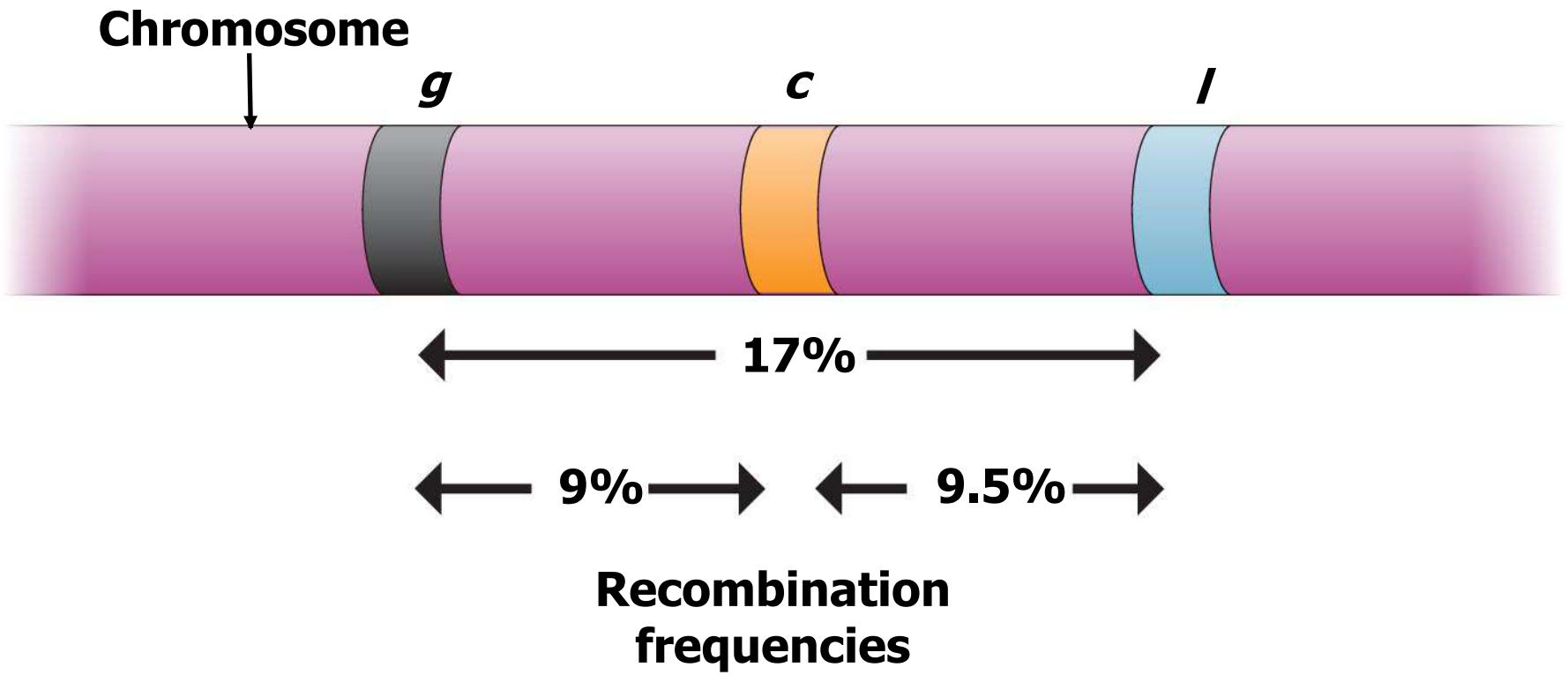


Figure 9.28

Linkage Maps

- Early studies of crossing over were performed using the fruit fly *Drosophila melanogaster*.
- Alfred H. Sturtevant, a student of Morgan, developed a method for mapping gene loci, which resulted in the creation of linkage maps.
  - A diagram of relative gene locations on a chromosome is a **linkage map**.

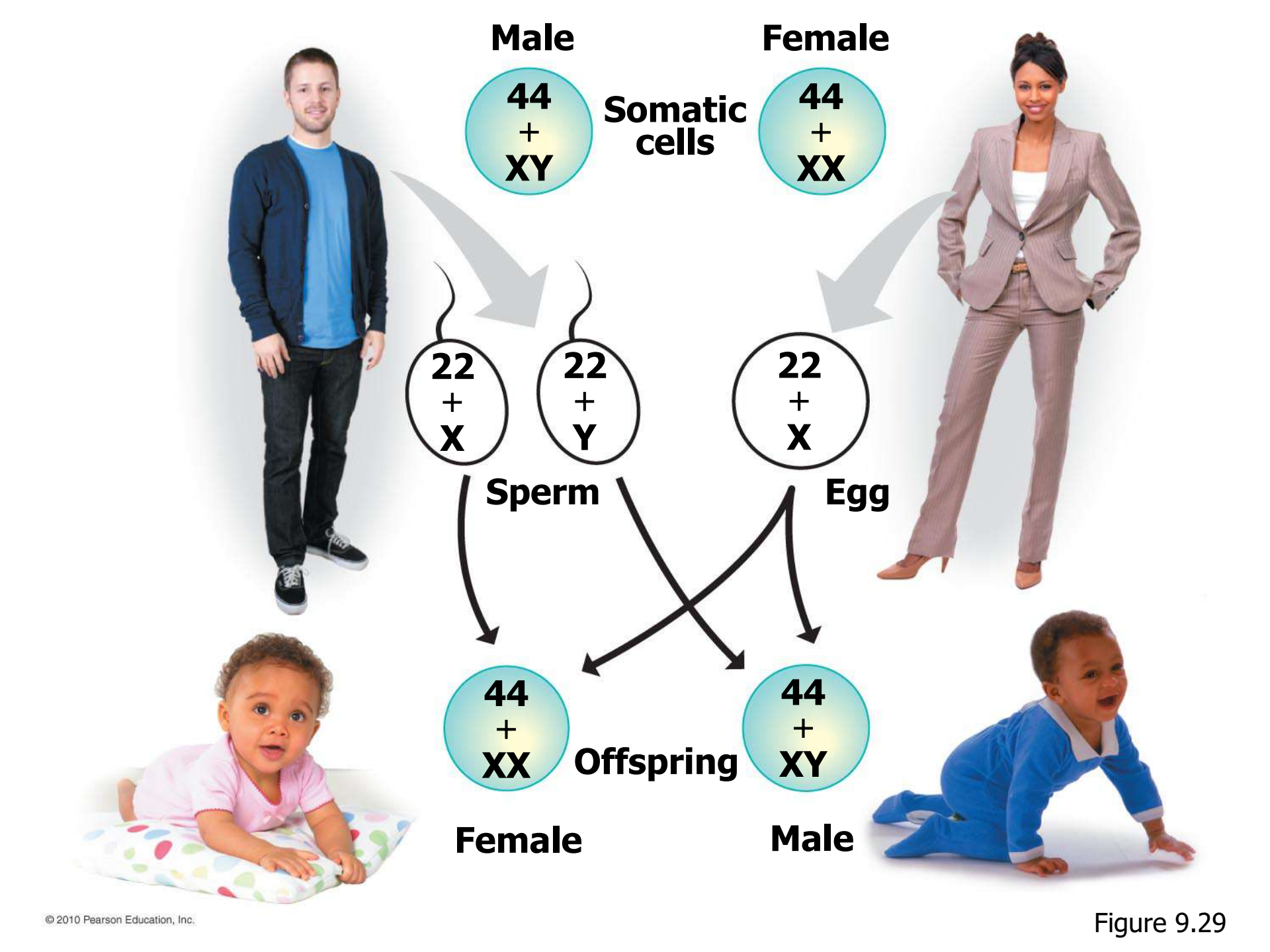
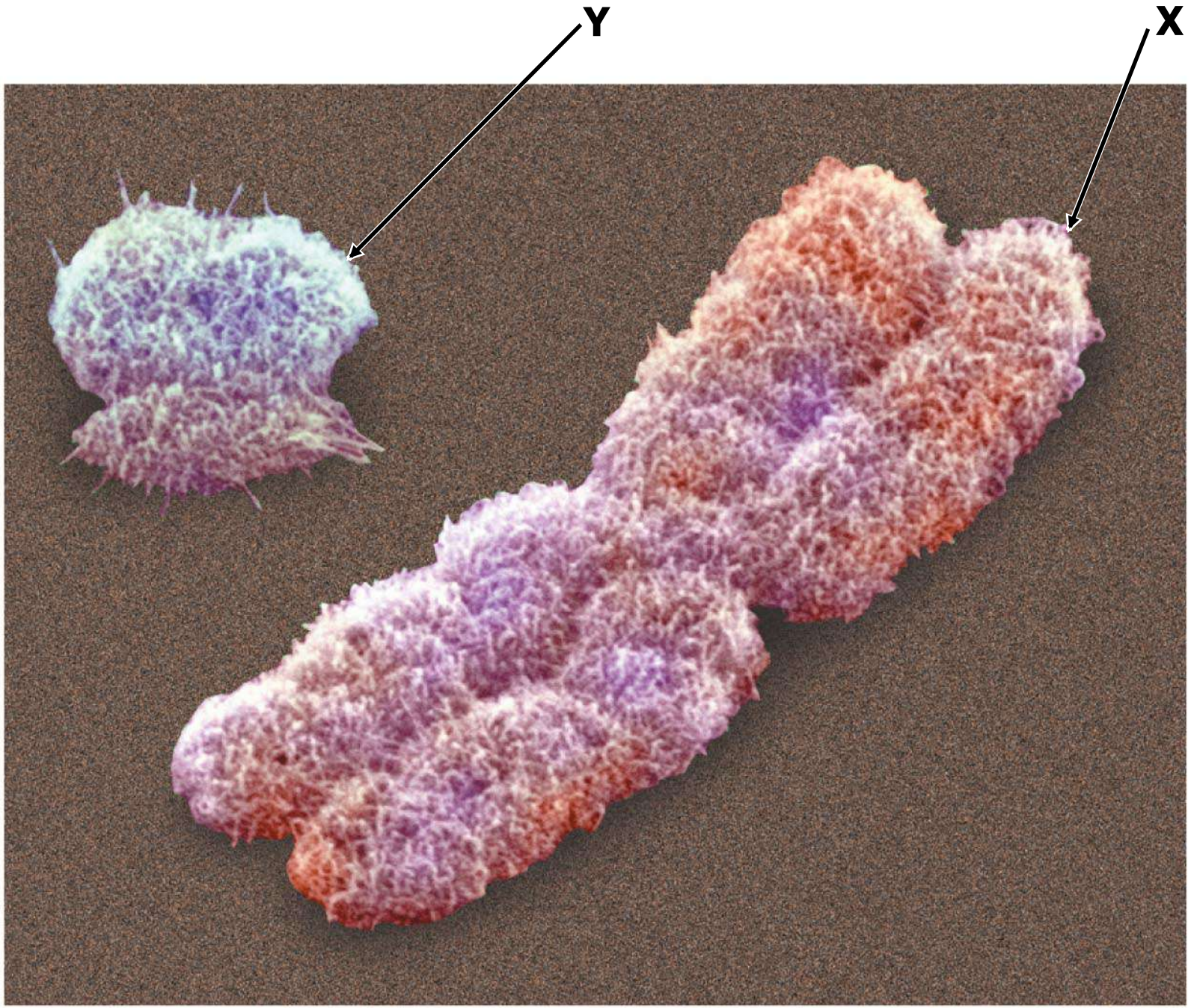


Figure 9.29



Colorized SEM

Figure 9.29

SEX CHROMOSOMES AND SEX-LINKED GENES

- Sex chromosomes influence the inheritance of certain traits.

Sex Determination in Humans

- Nearly all mammals have a pair of sex chromosomes designated X and Y.
  - Males have an X and Y.
  - Females have XX.

Sex-Linked Genes

- Any gene located on a sex chromosome is called a **sex-linked gene**.
  - Most sex-linked genes are found on the X chromosome.
  - **Red-green color blindness** is a common human sex-linked disorder.

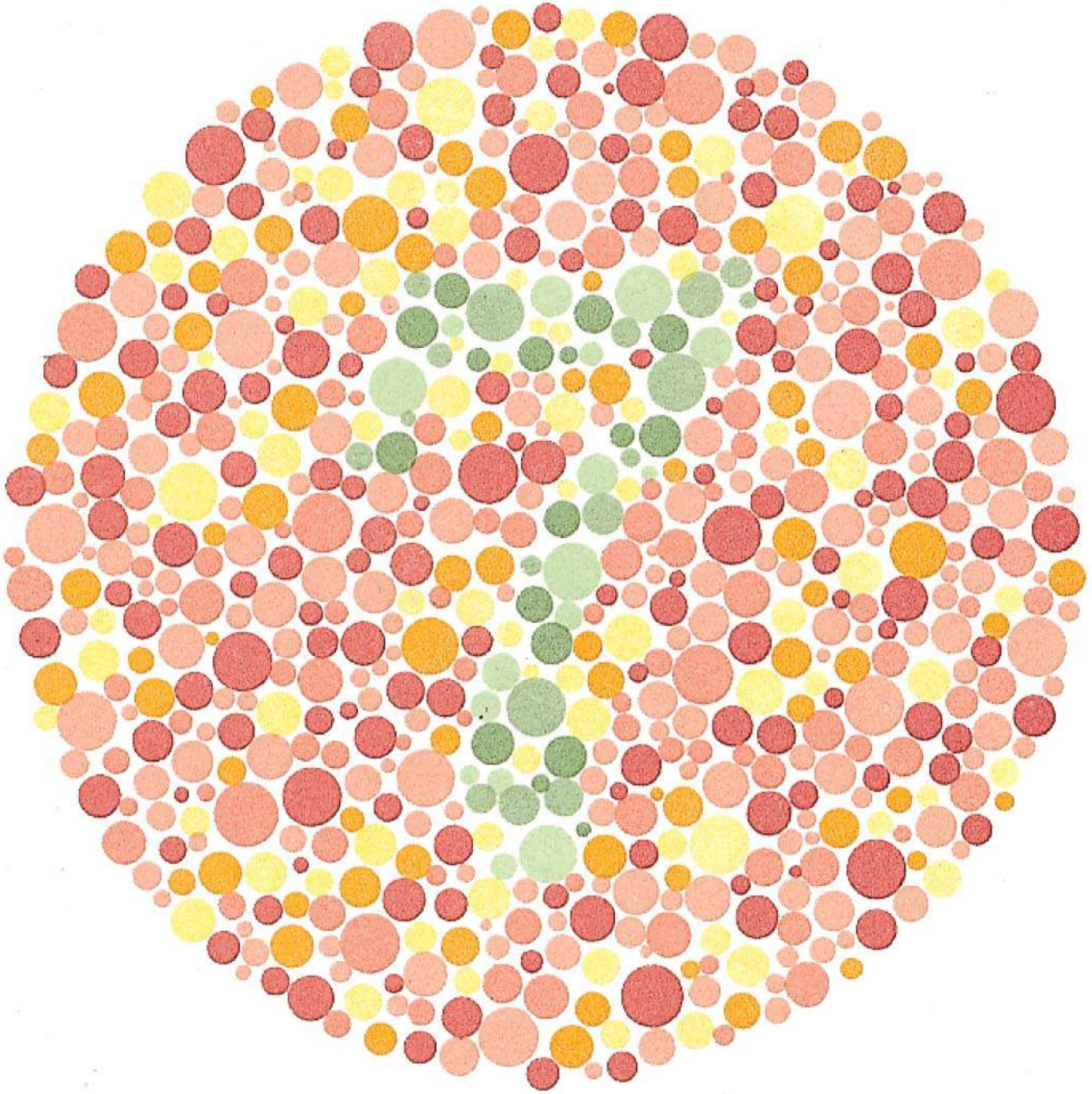


Figure 9.30

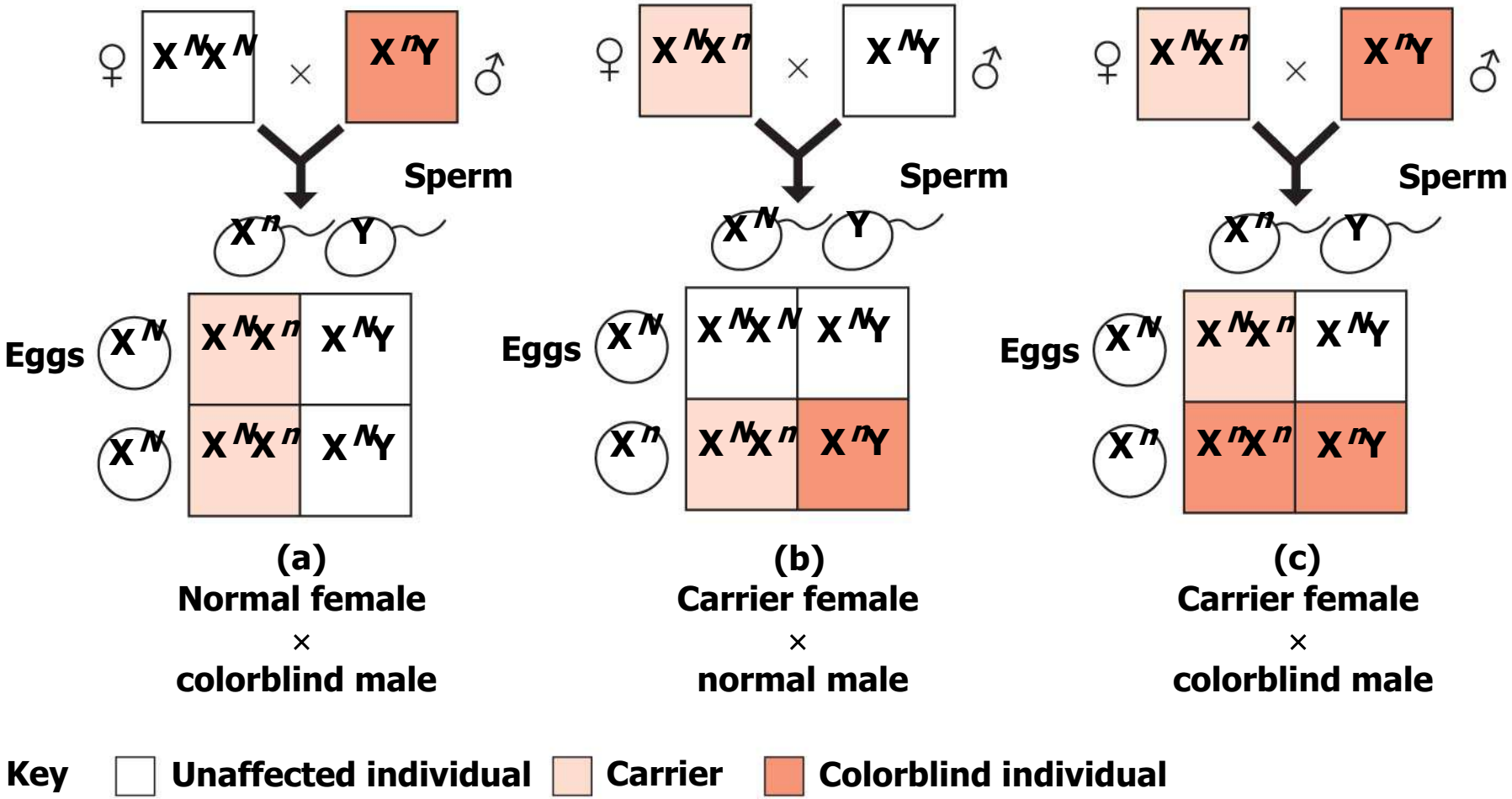


Figure 9.31

- **Hemophilia**
  - Is a sex-linked recessive blood-clotting trait that may result in excessive bleeding and death after relatively minor cuts and bruises
  - Has plagued royal families of Europe

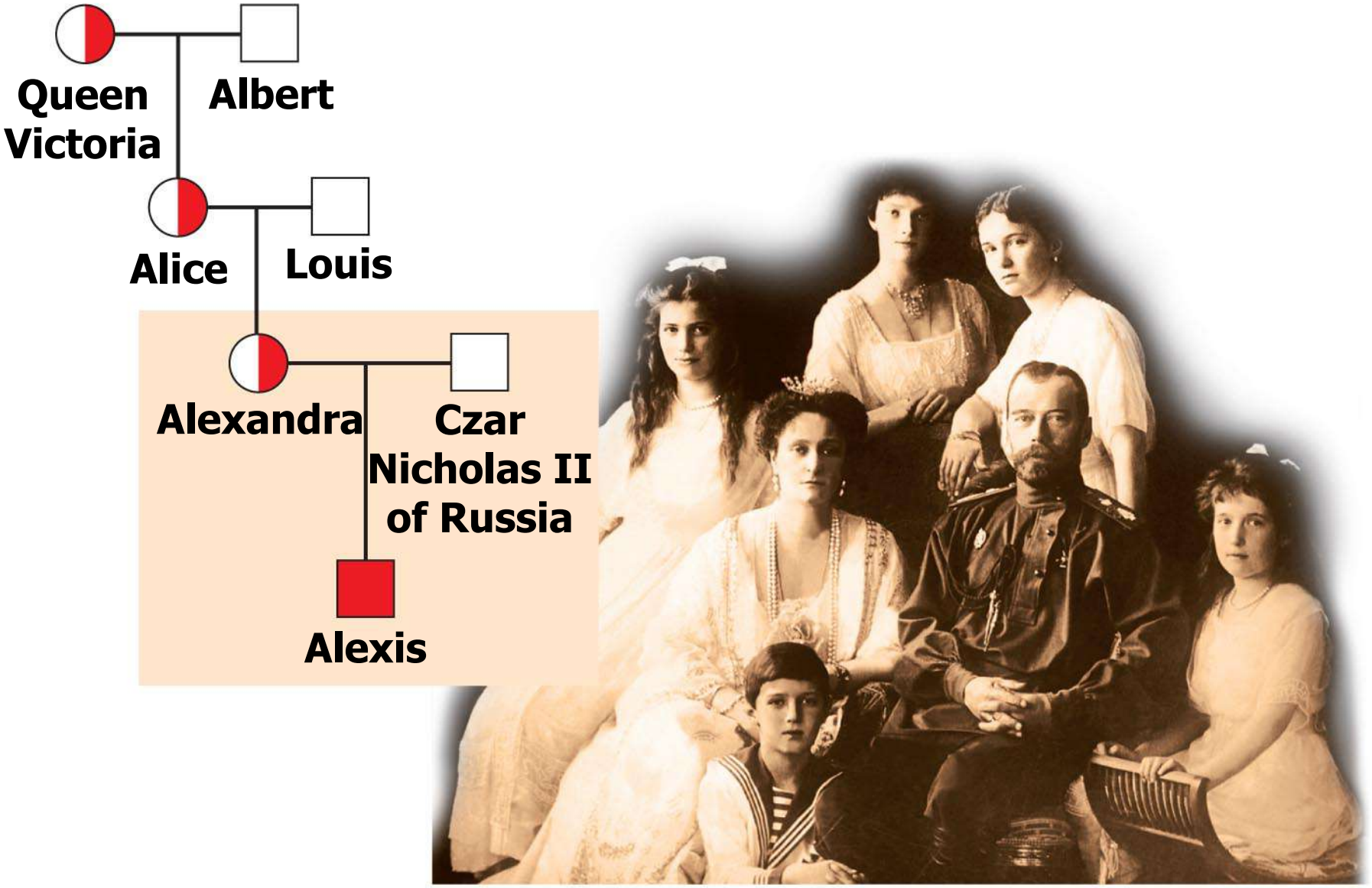


Figure 9.32

Evolution Connection: Barking Up the Evolutionary Tree

- About 15,000 years ago in East Asia, humans began to cohabit with ancestral canines that were predecessors of modern wolves and dogs.
- As people settled into geographically distinct populations, different canines became separated and inbred.

- In 2005 researchers sequenced the complete genome of a dog.
- An evolutionary tree of dog breeds was created.

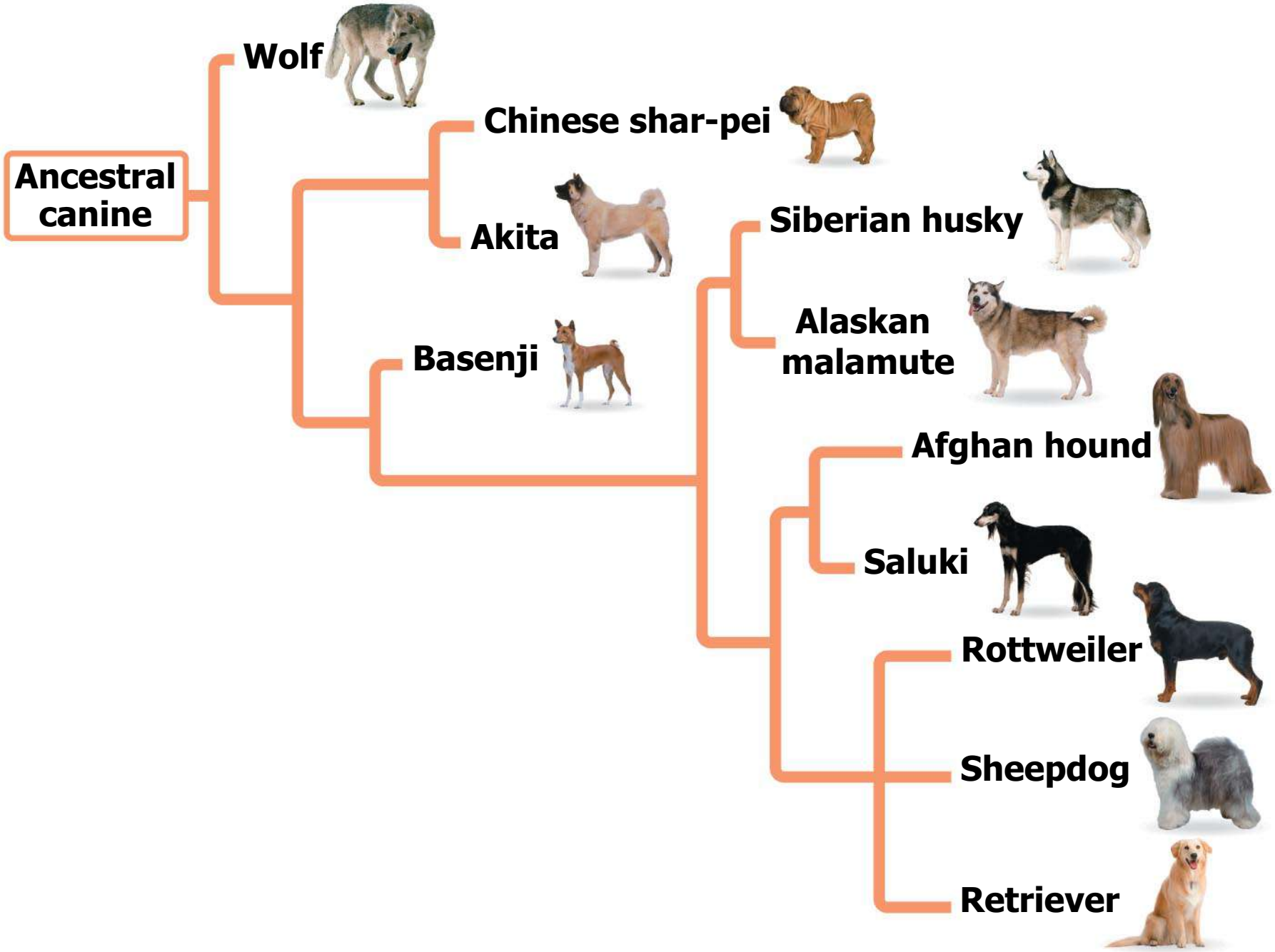


Figure 9.33

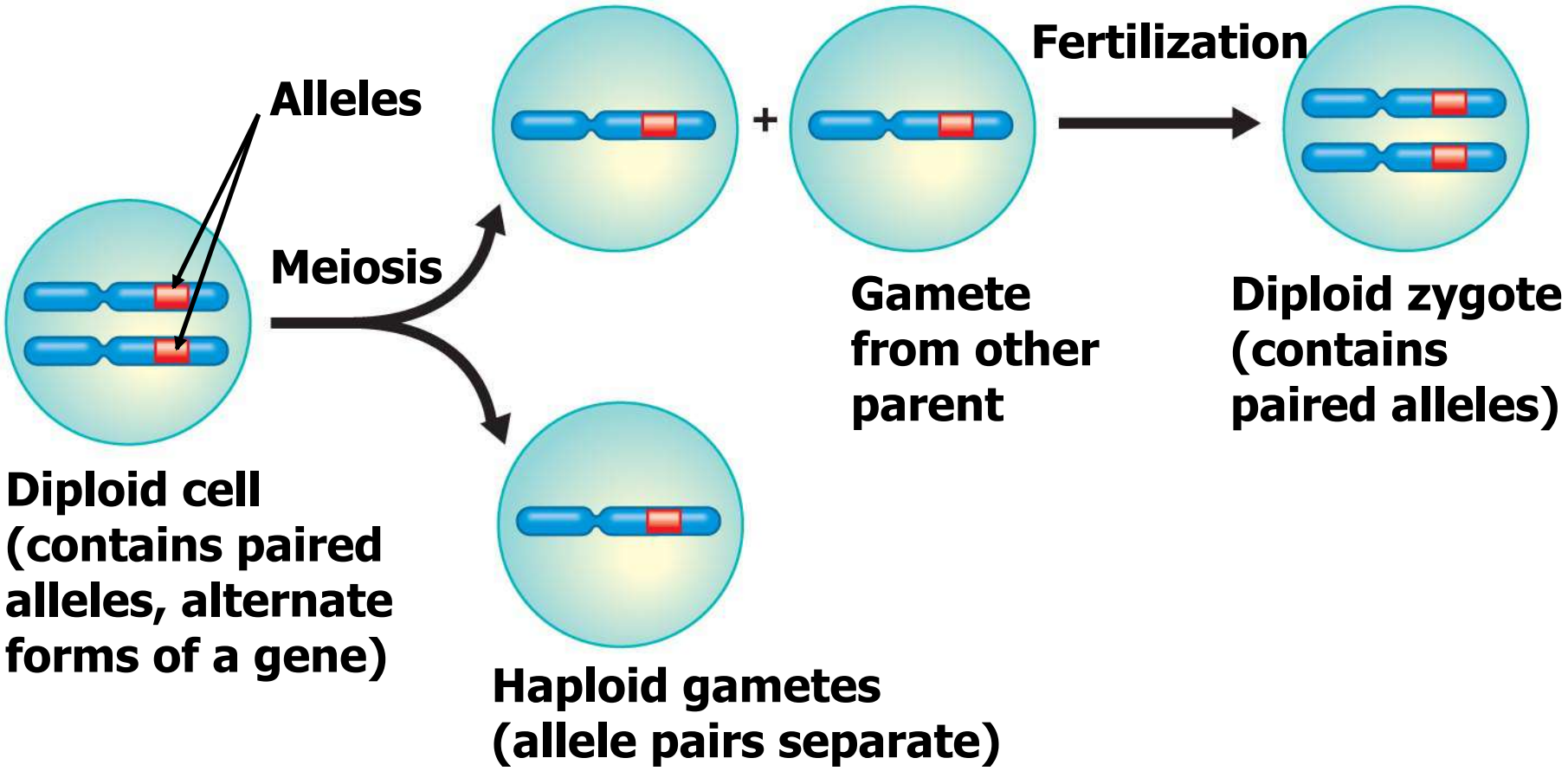


Figure 9.UN1

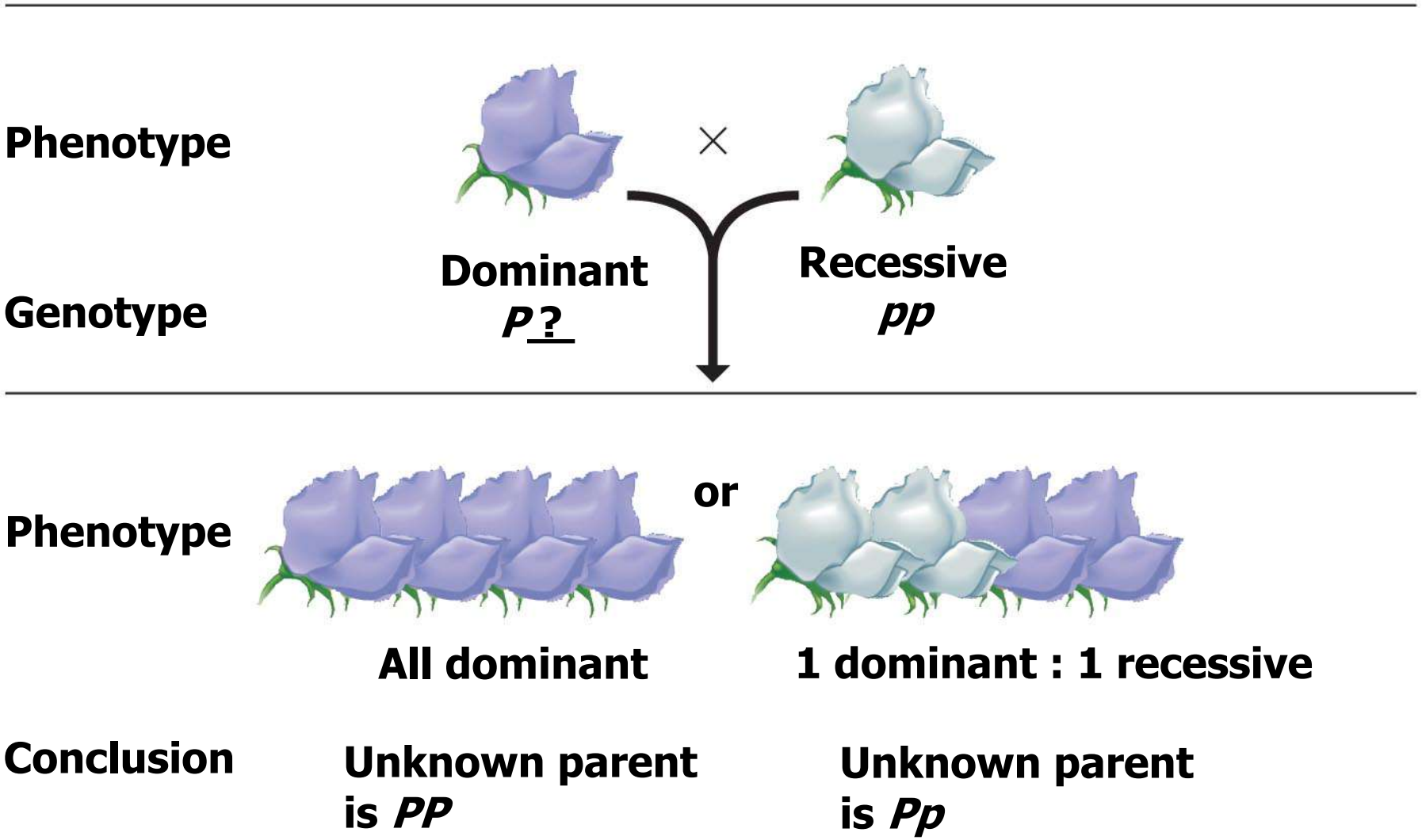


Figure 9.UN2

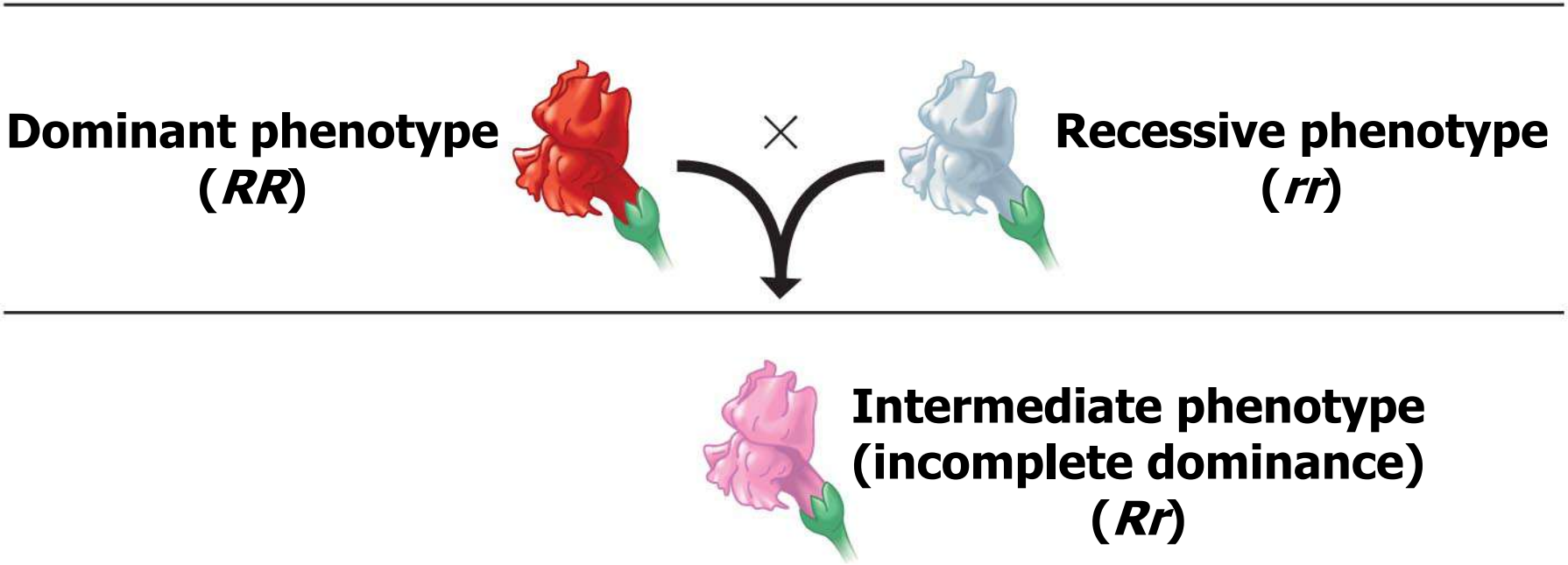


Figure 9.UN3

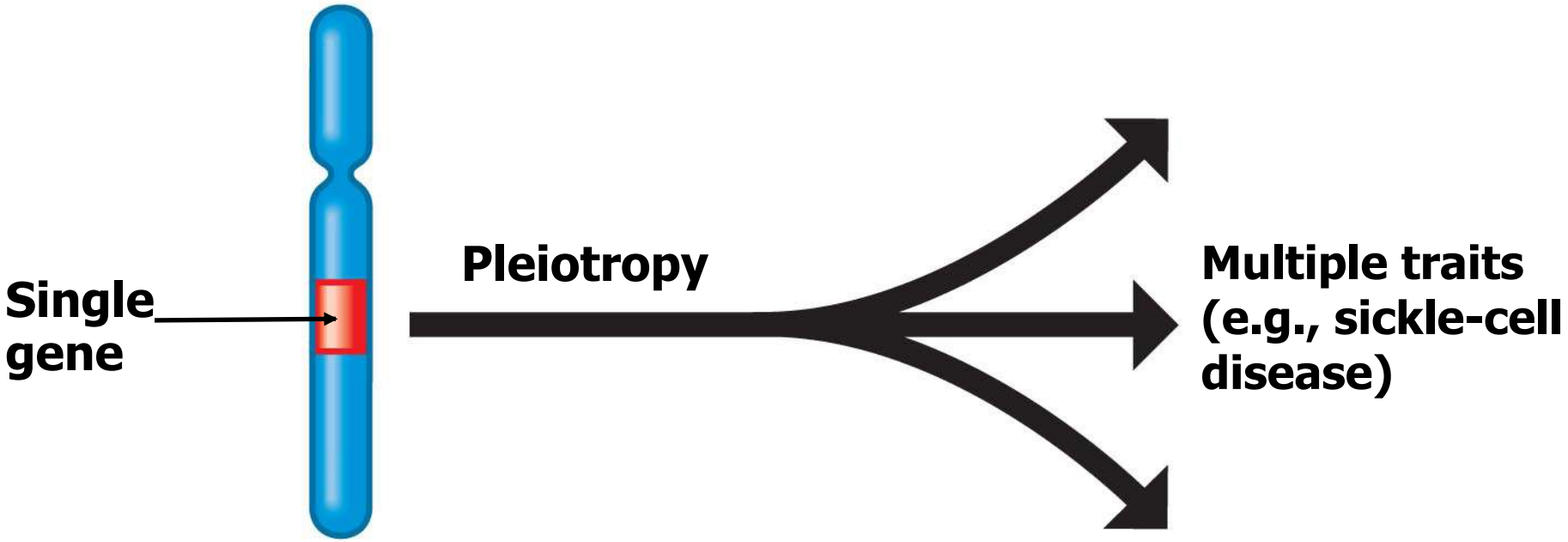


Figure 9.UN4

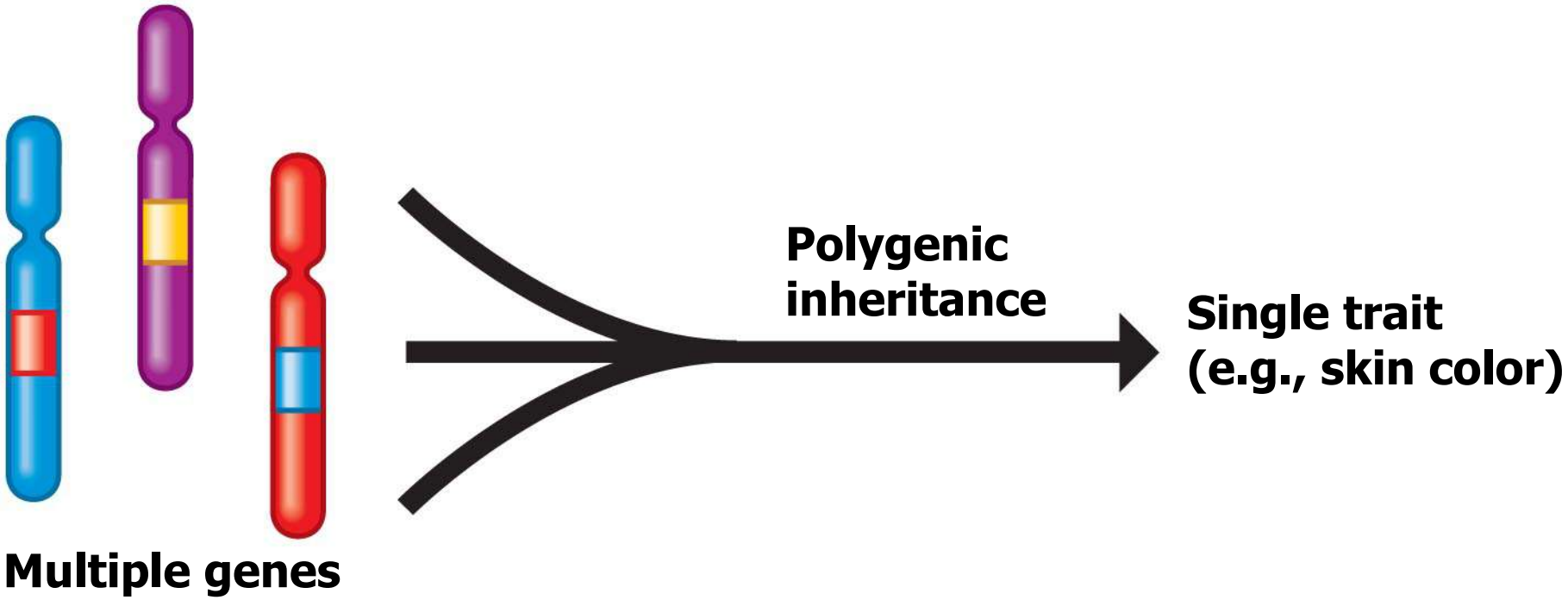


Figure 9.UN5

Male

44
+
XY

Somatic cells

Female

44
+
XX

Figure 9.UN6

Sex-Linked Traits				
Female: Two alleles	Genotype	$X^N X^N$	$X^N X^n$	$X^n X^n$
	Phenotype	Normal female	Carrier female	Affected female (rare)
Male: One allele	Genotype	$X^N Y$		$X^n Y$
	Phenotype	Normal male		Affected male

Figure 9.UN7