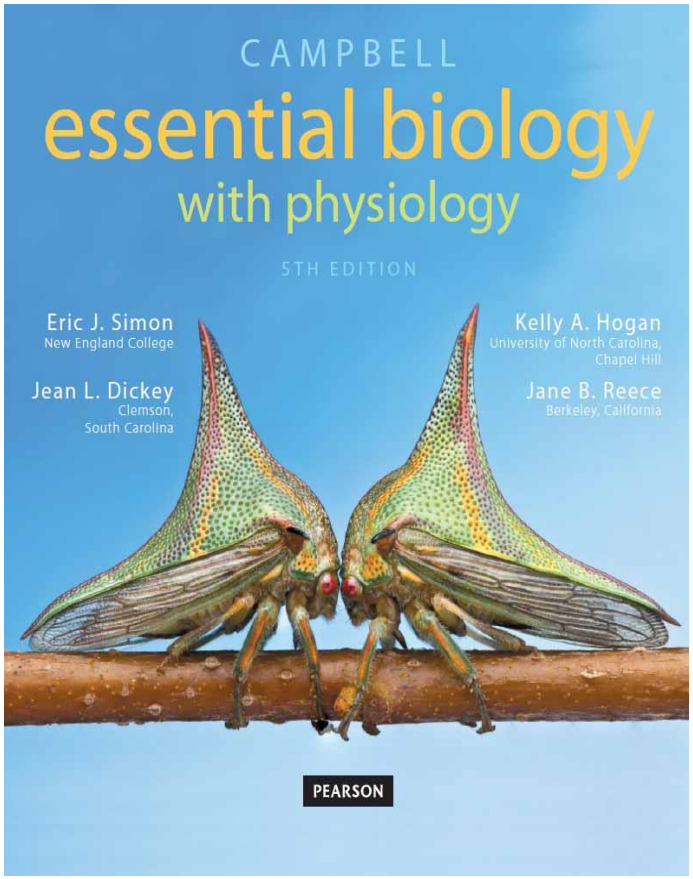
Essential Biology with Physiology

Fifth Edition



Chapter 9

Patterns of Inheritance

Pearson

Figure 9.0-1

Why Genetics Matters

Figure 9.0-1a

Figure 9.0-1b

Figure 9.0-1c

Biology and Society: Our Longest-Running Genetic Experiment

- People have selected and mated dogs with preferred traits for more than 15,000 years.
- Over thousands of years, such genetic tinkering has led to the incredible variety of body types and behaviors in dogs today.
- The biological principles underlying genetics have only recently been understood.

Figure 9.0 Breeding a Best Friend

Chapter Thread: Dog Breeding

Genetics and Heredity

- **Heredity** is the transmission of traits from one generation to the next.
- **Genetics** is the scientific study of heredity.
- Gregor Mendel worked in the 1860s and argued that
  - parents pass on to their offspring discrete genes (which he termed “heritable factors”),
  - genes are responsible for inherited traits, and
  - genes retain their individual identities generation after generation, no matter how they are mixed up or temporarily masked.

Figure 9.1 Gregor Mendel

In an Abbey Garden (1 of 6)

- Mendel probably chose to study garden peas because they
  - were easy to grow and
  - came in many readily distinguishable varieties.

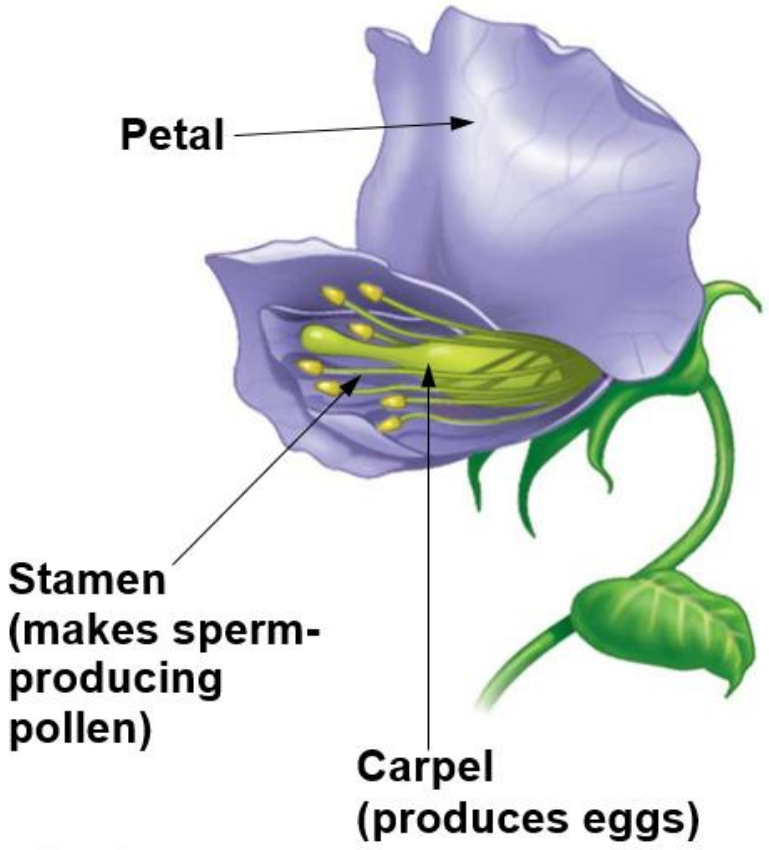
In an Abbey Garden (2 of 6)

- 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 traits.

In an Abbey Garden (3 of 6)

- Perhaps the most important advantage of pea plants as an experimental model was that Mendel could strictly control their reproduction.
  - The petals of a pea flower almost completely enclose the egg-producing organ (the carpel) and the sperm-producing organs (the stamens).
  - Consequently, pea plants usually self-fertilize because sperm-carrying pollen grains released from the stamens land on the tip of the egg-containing carpel of the same flower.

Figure 9.2 The Structure of a Pea Flower



In an Abbey Garden (4 of 6)

- When Mendel wanted to fertilize one plant with pollen from a different plant, he pollinated the plants by hand and was always sure of the parentage of his new plants.

Figure 9.3 Mendel's Technique for Cross-Fertilizing Pea Plants (1 of 3)

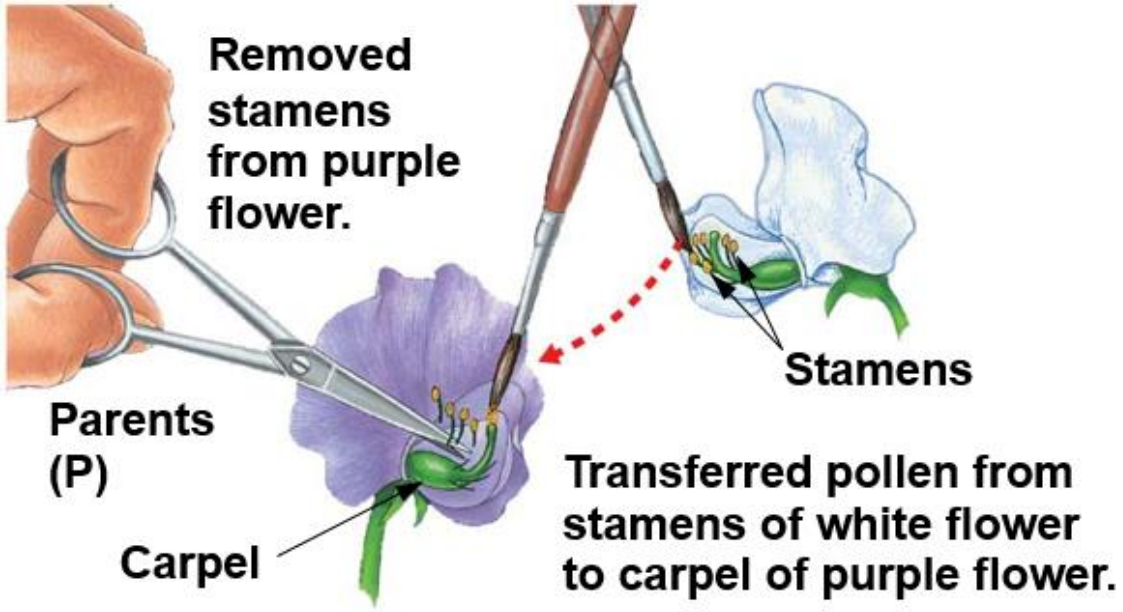


Figure 9.3 Mendel's Technique for Cross-Fertilizing Pea Plants (2 of 3)

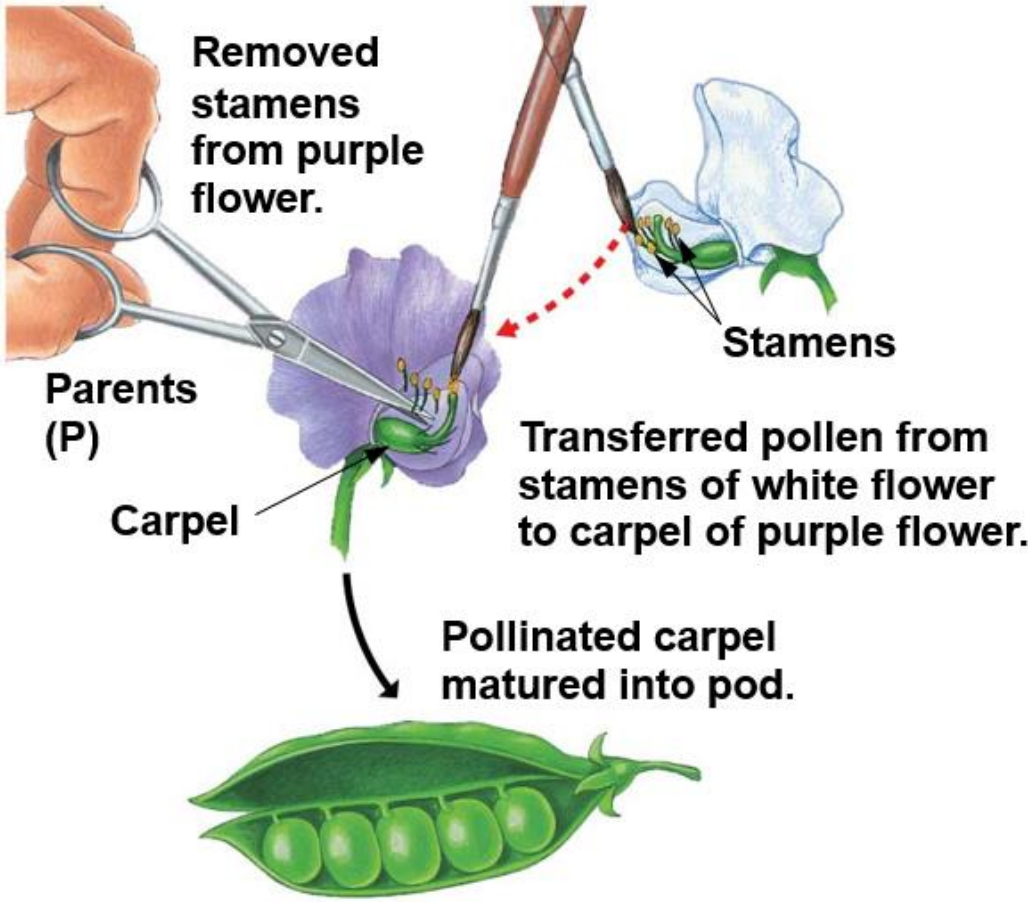
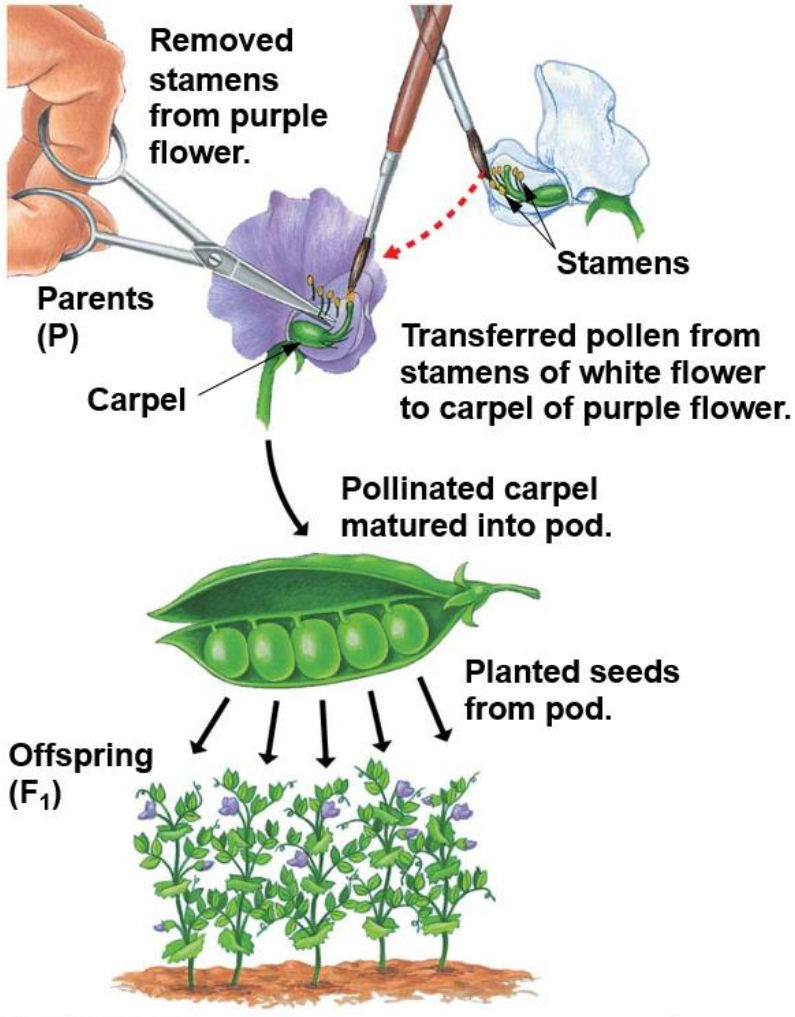


Figure 9.3 Mendel's Technique for Cross-Fertilizing Pea Plants (3 of 3)



In an Abbey Garden (5 of 6)

- Mendel
  - created purebred varieties of plants and
  - crossed two different purebred varieties.

In an Abbey Garden (6 of 6)

- **Hybrids** are the offspring of two different purebred varieties.
  - The cross-fertilization itself is referred to as a genetic **cross**.
  - 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**.

Mendel's Law of Segregation

- Mendel performed many experiments in which he tracked the inheritance of characters, such as flower color, that occur as two alternative traits.
- The results led him to formulate several hypotheses about inheritance.

Figure 9.4 The Seven Characters of Pea Plants Studied by Mendel (1 of 2)

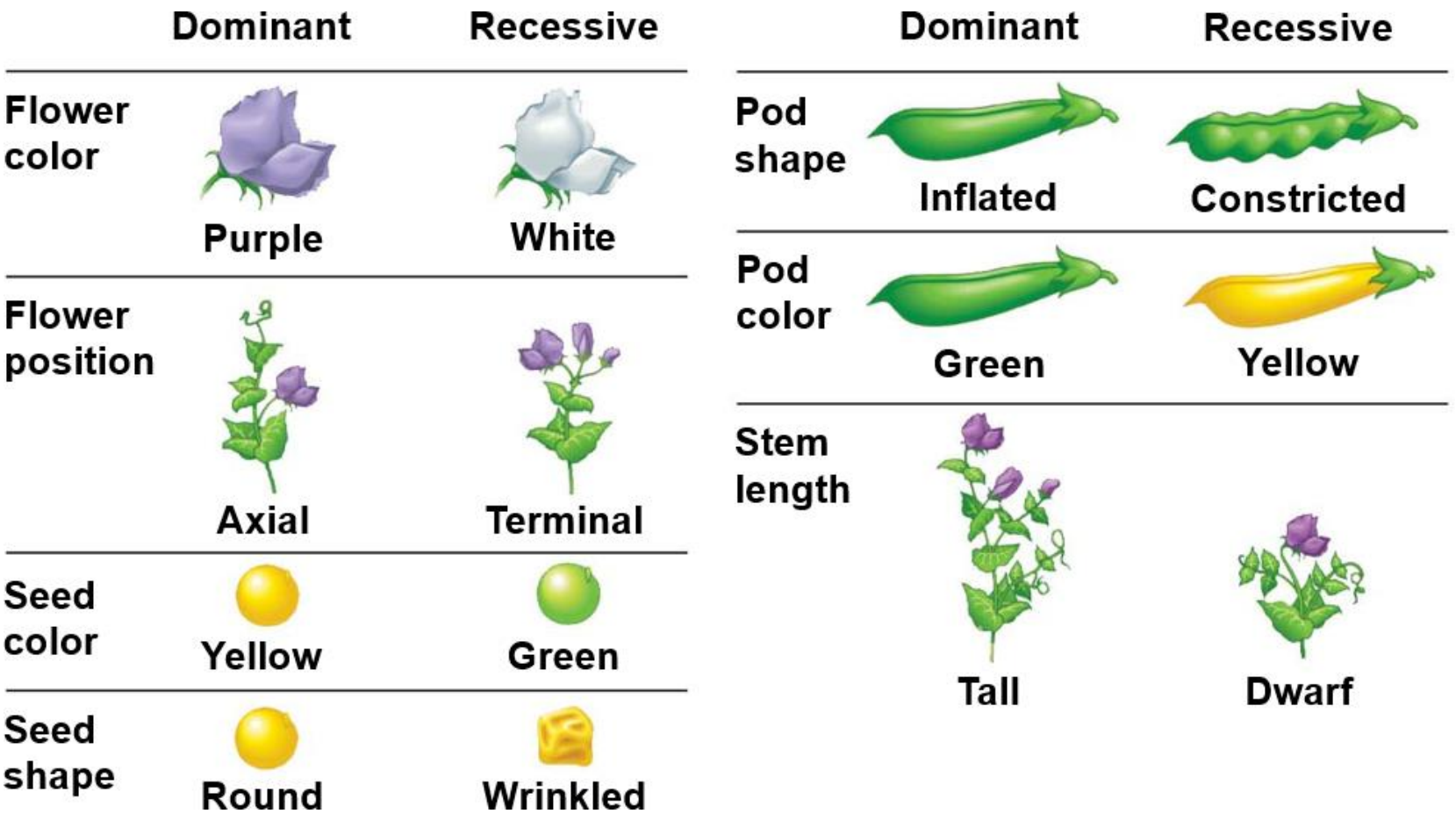


Figure 9.4 The Seven Characters of Pea Plants Studied by Mendel (2 of 2)

Monohybrid Crosses (1 of 9)

- Mendel performed a **monohybrid cross** between purebred parent plants that differ in only one character and found that the F_1 plants all had purple flowers.
  - Was the factor responsible for inheritance of white flowers now lost as a result of the cross?
  - By mating the F_1 plants with each other, Mendel found the answer to this question to be no.

Figure 9.5 Mendel's Cross Tracking One Character (Flower Color) (1 of 4)

Figure 9.5 Mendel's Cross Tracking One Character (Flower Color) (2 of 4)

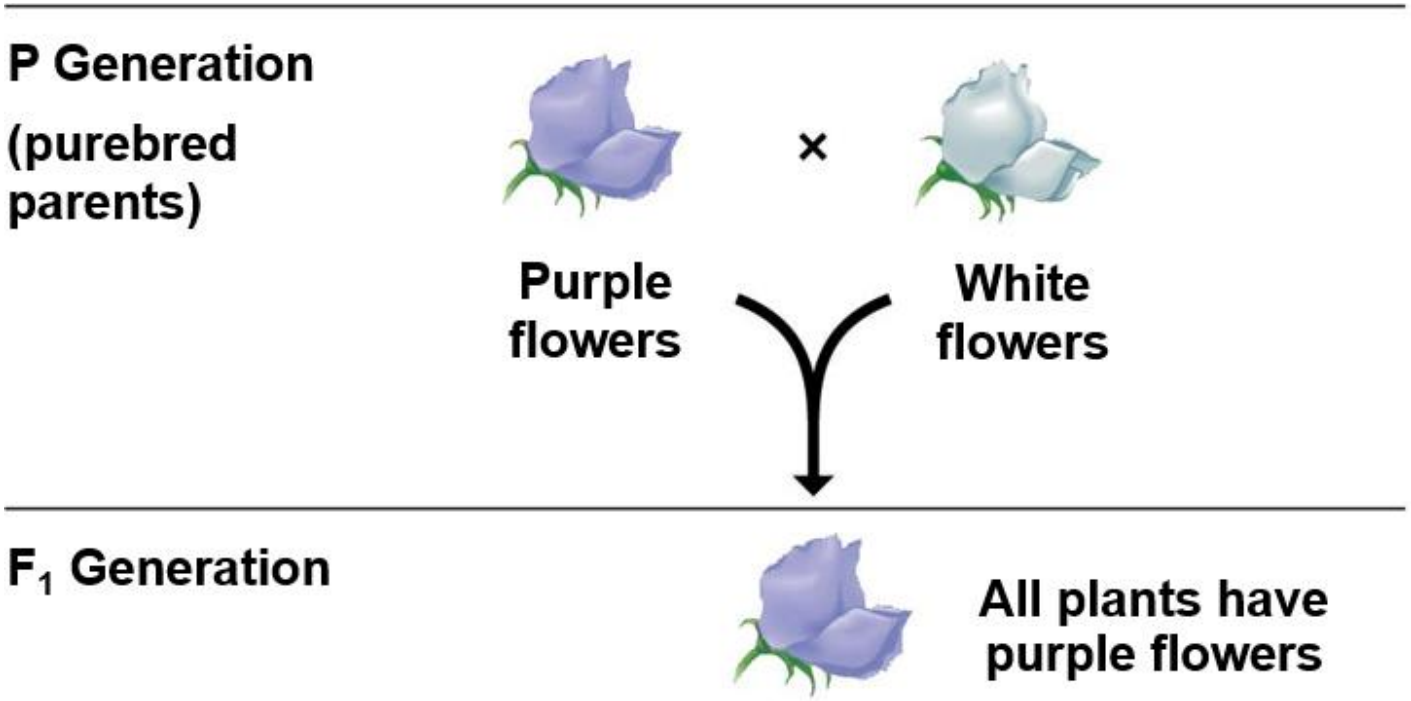


Figure 9.5 Mendel's Cross Tracking One Character (Flower Color) (3 of 4)

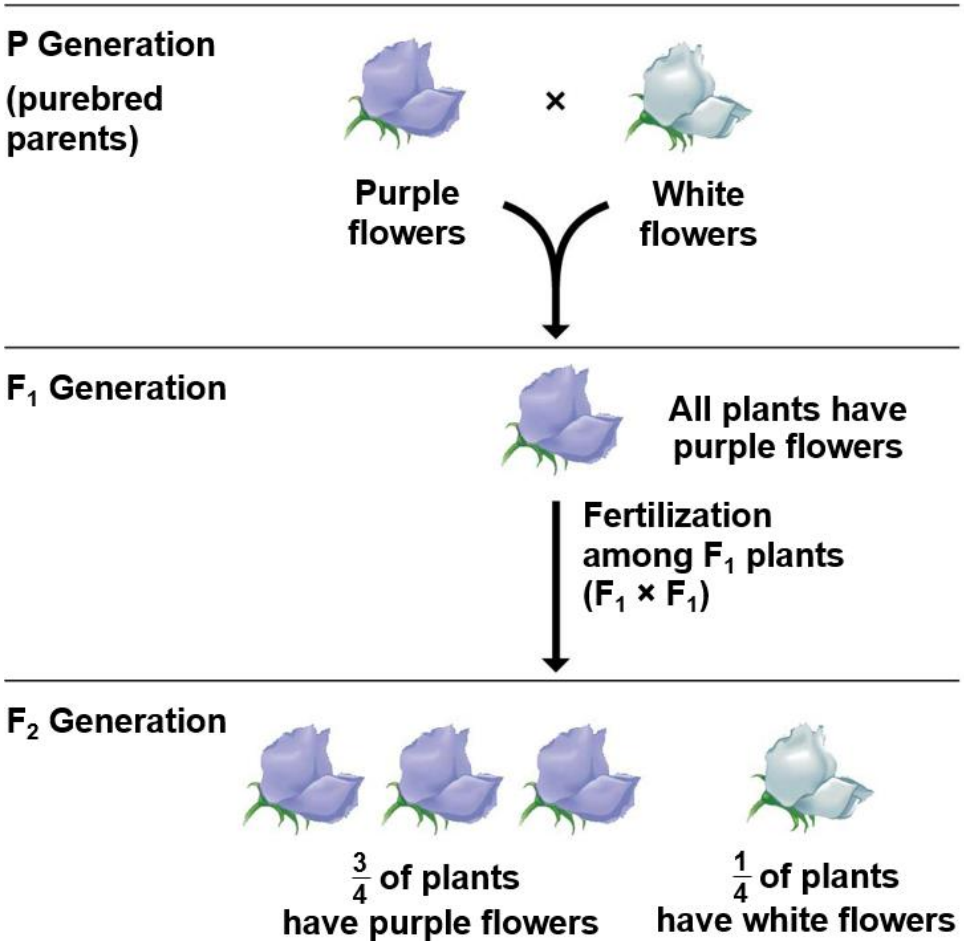


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Figure 9.5 Mendel's Cross Tracking One Character (Flower Color) (4 of 4)

Monohybrid Crosses (2 of 9)

- Mendel figured out that the gene for white flowers did not disappear in the F_1 plants but was somehow hidden or masked when the purple-flower factor was present.
- He also deduced that the F_1 plants must have carried two factors for the flower-color character,
  - one for purple and
  - one for white.

Monohybrid Crosses (3 of 9)

- From these results and others, Mendel developed four hypotheses:
  1. **There are alternative versions of genes that account for variations in inherited characters.**
     - The alternative versions of genes are called **alleles**.

Monohybrid Crosses (4 of 9)

2. **For each inherited character, an organism inherits two alleles, one from each parent.**
   - An organism that has two identical alleles for a gene is said to be **homozygous** for that gene.
   - An organism that has two different alleles for a gene is said to be **heterozygous** for that gene.

Monohybrid Crosses (5 of 9)

3. **If the two alleles of an inherited pair differ, then one determines the organism's appearance and is called the dominant allele, and the other has no noticeable effect on the organism's appearance and is called the recessive allele.**
   - Geneticists use uppercase italic letters (such as *P*) to represent dominant alleles and lowercase italic letters (such as *p*) to represent recessive alleles.

Monohybrid Crosses (6 of 9)

4. **A sperm or egg carries only one allele for each inherited character because the two alleles for a character segregate (separate) from each other during the production of gametes.**
   - This statement is called the **law of segregation**.
   - When sperm and egg unite at fertilization, each contributes its alleles, restoring the paired condition in the offspring.
   - **Figure 9.6** illustrates Mendel's law of segregation, which explains the inheritance pattern shown in Figure 9.5.

Figure 9.6 The Law of Segregation (1 of 3)

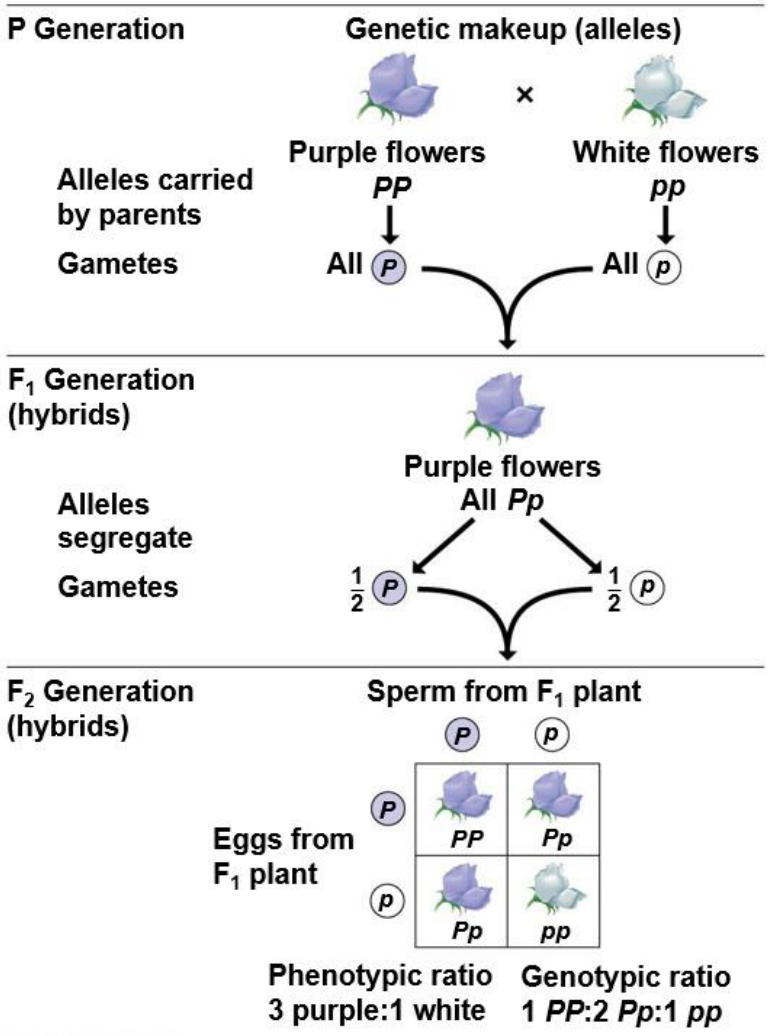


Figure 9.6 The Law of Segregation (2 of 3)

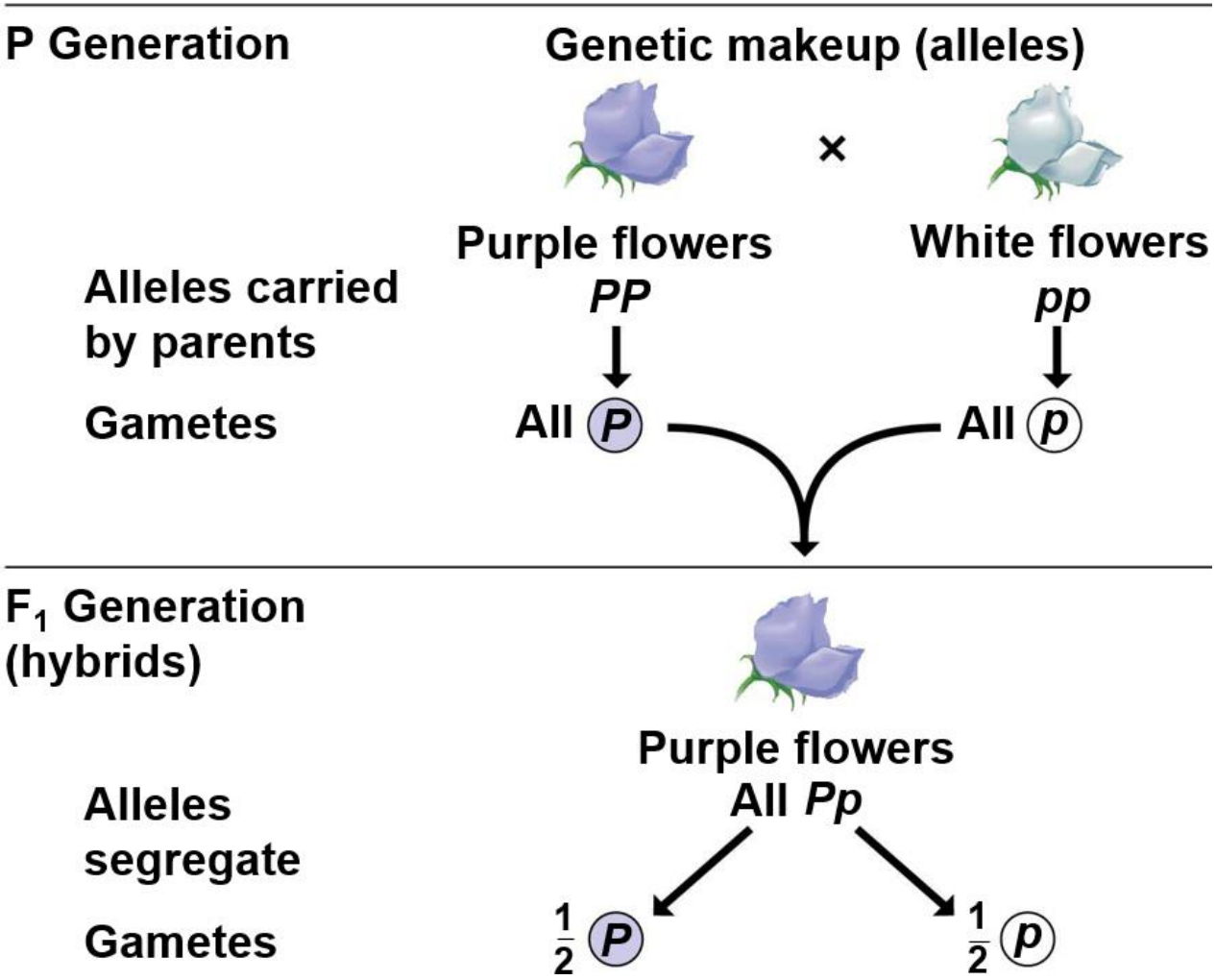
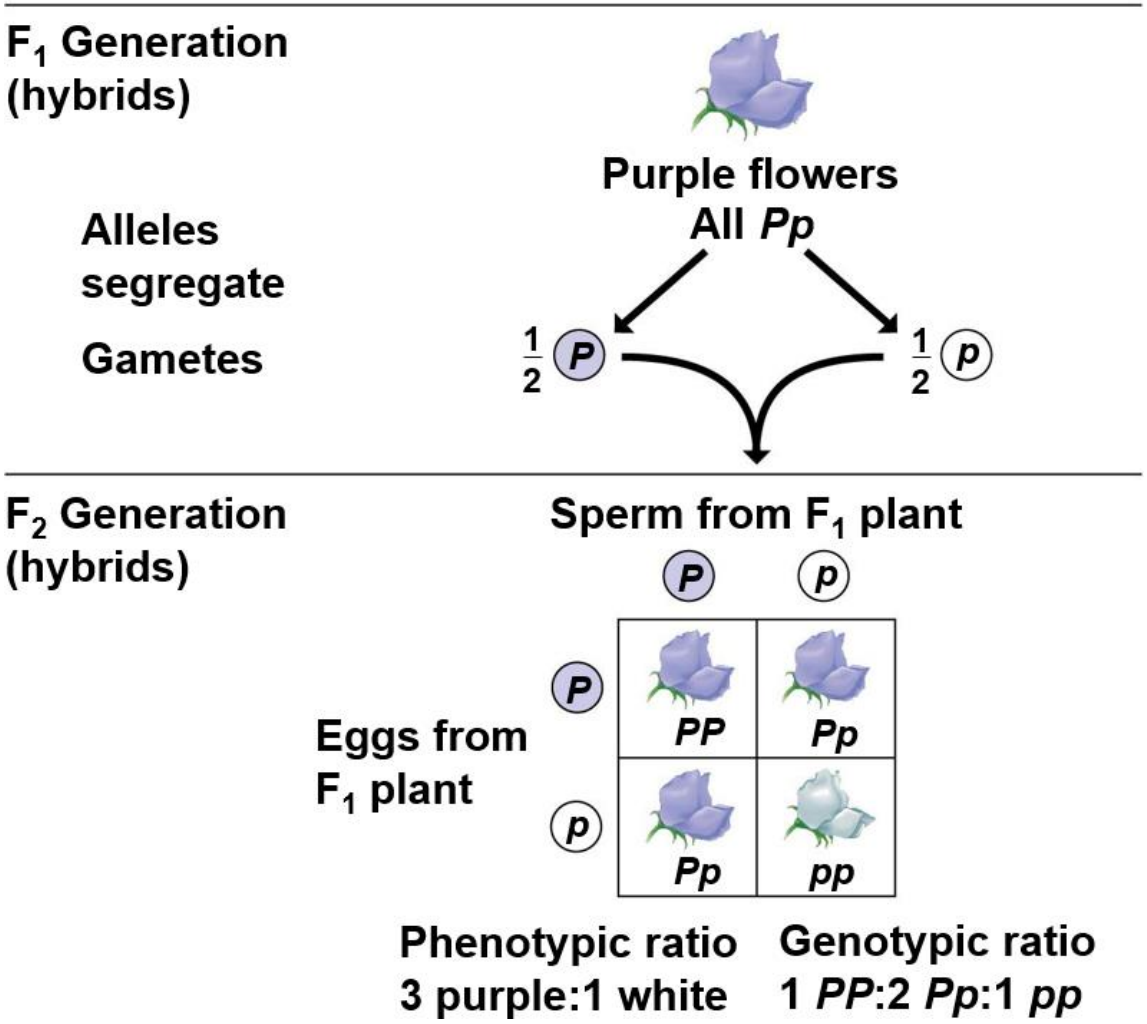


Figure 9.6 The Law of Segregation (3 of 3)



Monohybrid Crosses (7 of 9)

- A **Punnett square** highlights
  - the four possible combinations of gametes and
  - the resulting four possible offspring in the F_2 generation.
- Each square represents an equally probable product of fertilization.

Monohybrid Crosses (8 of 9)

- Geneticists distinguish between an organism's
  - physical appearance, its **phenotype**, and
  - genetic makeup, its **genotype**.

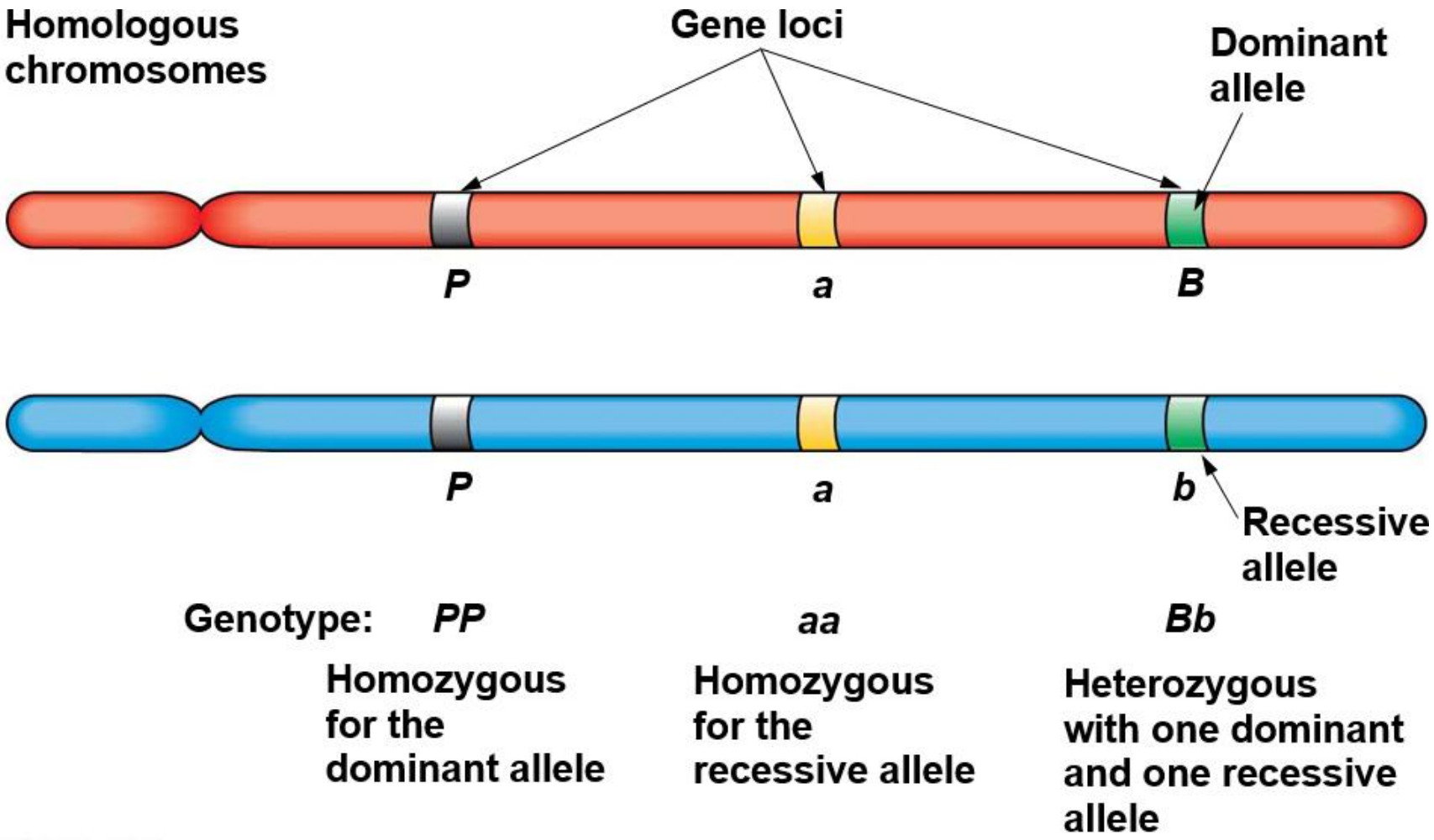
Monohybrid Crosses (9 of 9)

- Mendel found that each of the seven characters he studied had the same inheritance pattern: A parental trait disappeared in the F_1 generation, only to reappear in one-fourth of the F_2 offspring.
  - The underlying mechanism is explained by Mendel's law of segregation:
    - Pairs of alleles segregate during gamete formation;
    - the fusion of gametes at fertilization creates allele pairs again.

Genetic Alleles and Homologous Chromosomes

- The diagram in **Figure 9.7** shows a pair of homologous chromosomes—chromosomes that carry alleles of the same genes.
- A gene **locus** is a specific location of a gene along a chromosome.
  - Alleles (alternative versions) of a gene reside at the same locus on homologous chromosomes.
  - However, the two chromosomes may bear either identical alleles or different ones at any one locus.

Figure 9.7 The Relationship Between Alleles and Homologous Chromosomes



Mendel's Law of Independent Assortment (1 of 4)

- A **dihybrid cross** is the mating of parental varieties differing in two characters.
- What would result from a dihybrid cross?

Mendel's Law of Independent Assortment (2 of 4)

- If the genes for the two characters were inherited together, then
  - the F_1 hybrids would produce only the same two kinds of gametes that they received from their parents, and
  - the F_2 generation would show a 3:1 phenotypic ratio.

Mendel's Law of Independent Assortment (3 of 4)

- If, however, the two seed characters sorted independently, then
  - the F_1 generation would produce four gamete genotypes (**RY**, **rY**, **Ry**, and **ry**) in equal quantities, and
  - the F_2 generation would have nine different genotypes producing four different phenotypes in a ratio of 9:3:3:1.

Figure 9.8 Testing Alternative Hypotheses for Gene Assortment in a Dihybrid Cross (1 of 5)

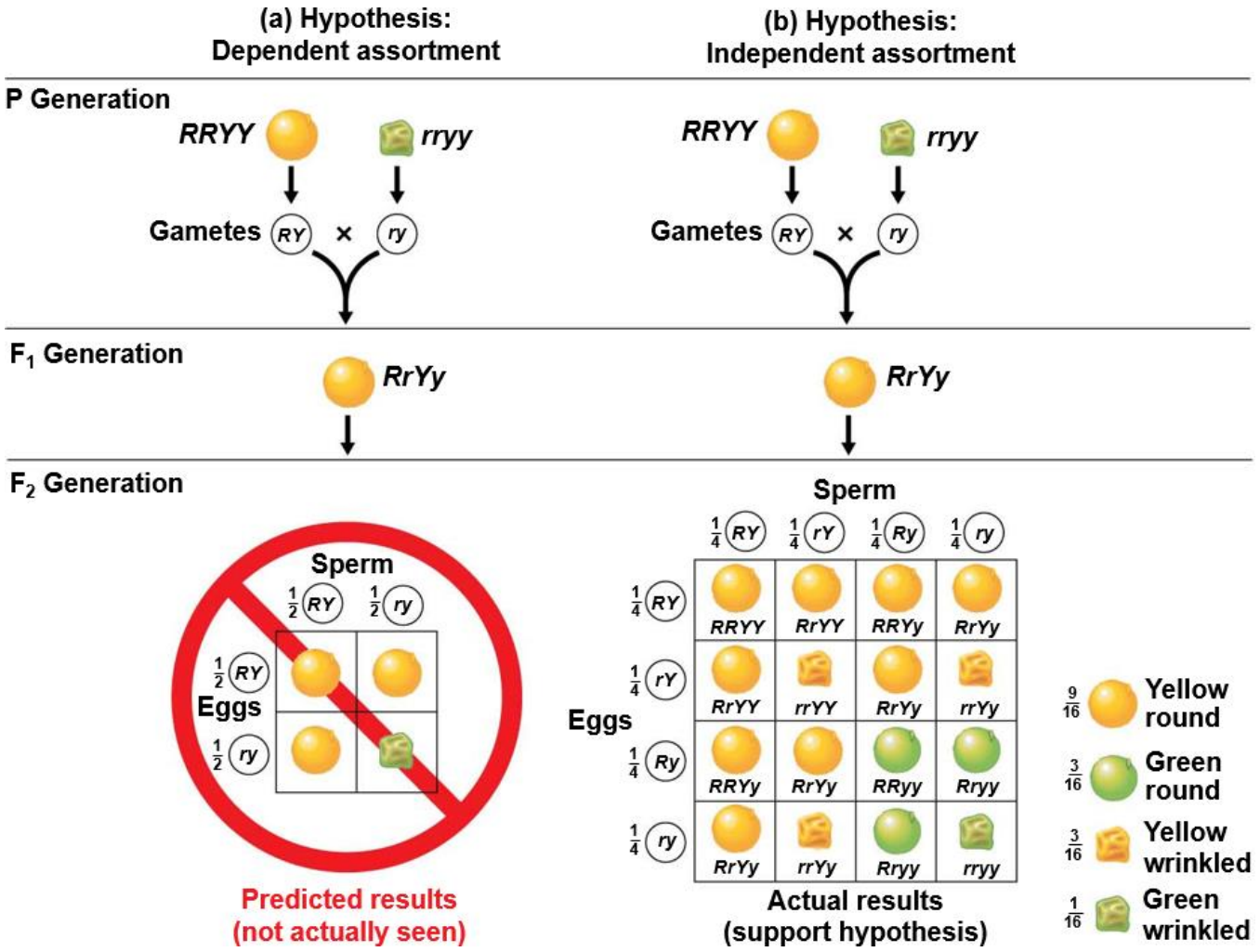


Figure 9.8 Testing Alternative Hypotheses for Gene Assortment in a Dihybrid Cross (2 of 5)

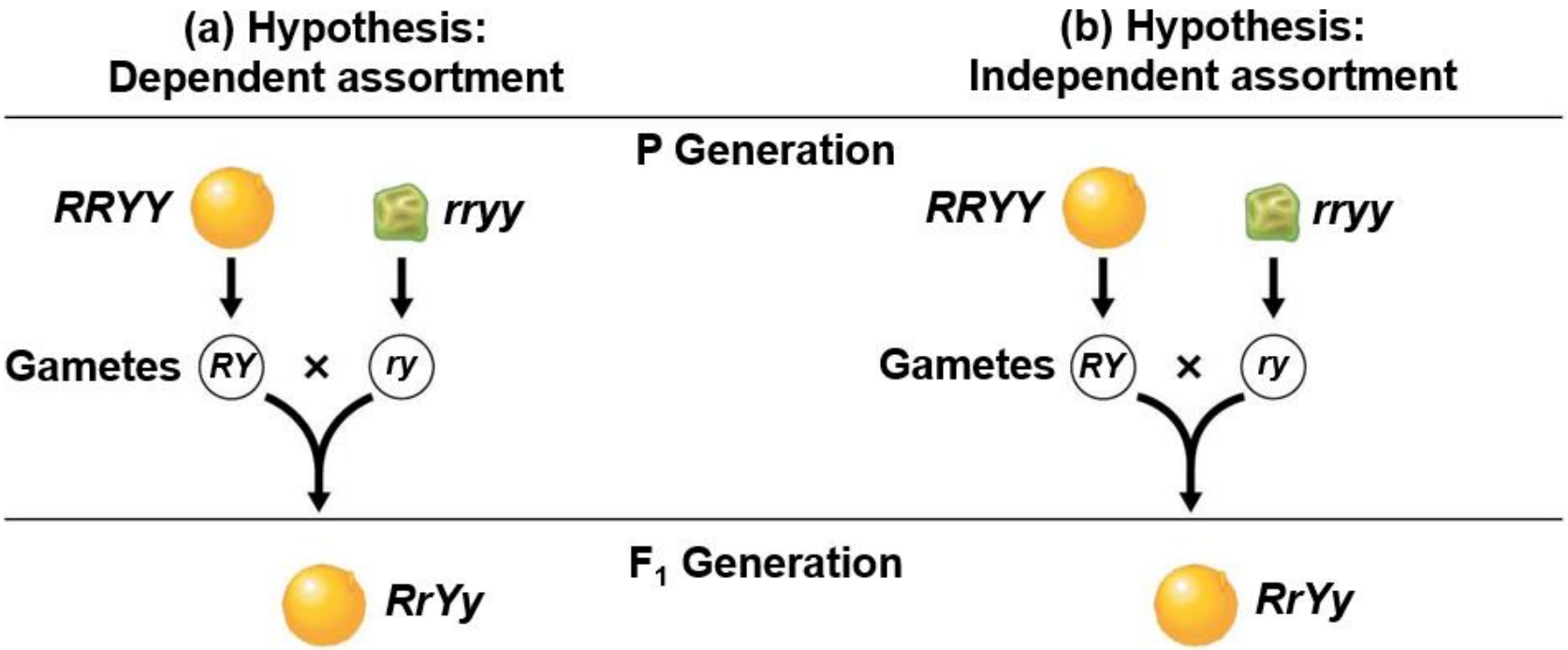


Figure 9.8 Testing Alternative Hypotheses for Gene Assortment in a Dihybrid Cross (3 of 5)

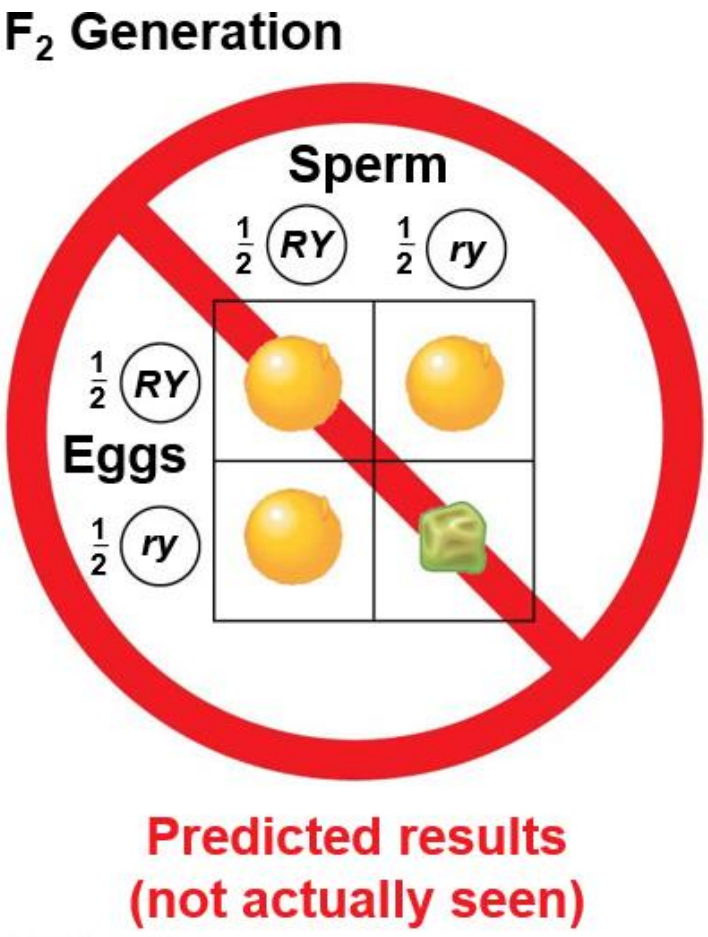


Figure 9.8 Testing Alternative Hypotheses for Gene Assortment in a Dihybrid Cross (4 of 5)

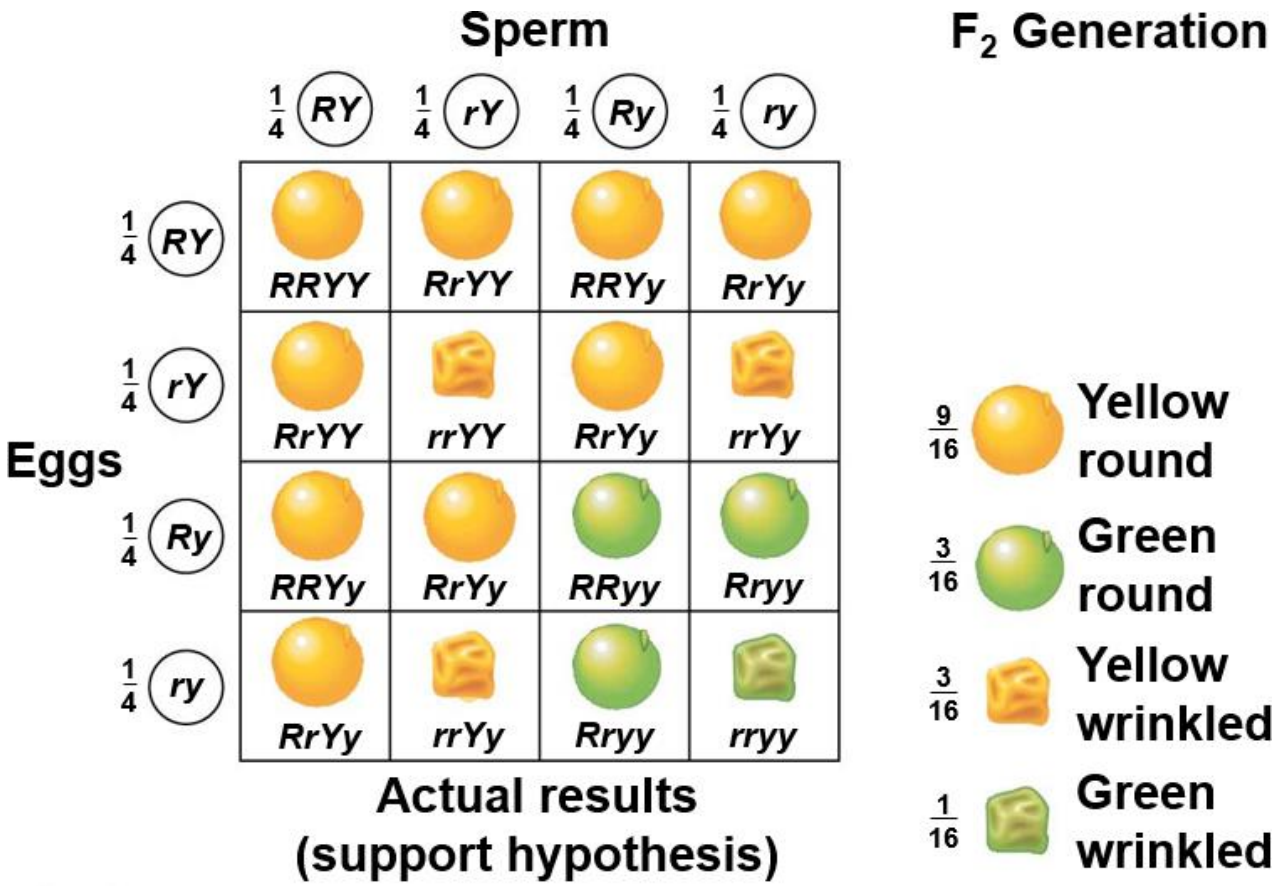


Figure 9.8 Testing Alternative Hypotheses for Gene Assortment in a Dihybrid Cross (5 of 5)

Mendel's Law of Independent Assortment (4 of 4)

- 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 called Mendel's **law of independent assortment**.
- Independent assortment is seen in the inheritance of two characters in Labrador retrievers.

Figure 9.9 Independent Assortment of Genes in Labrador Retrievers (1 of 3)

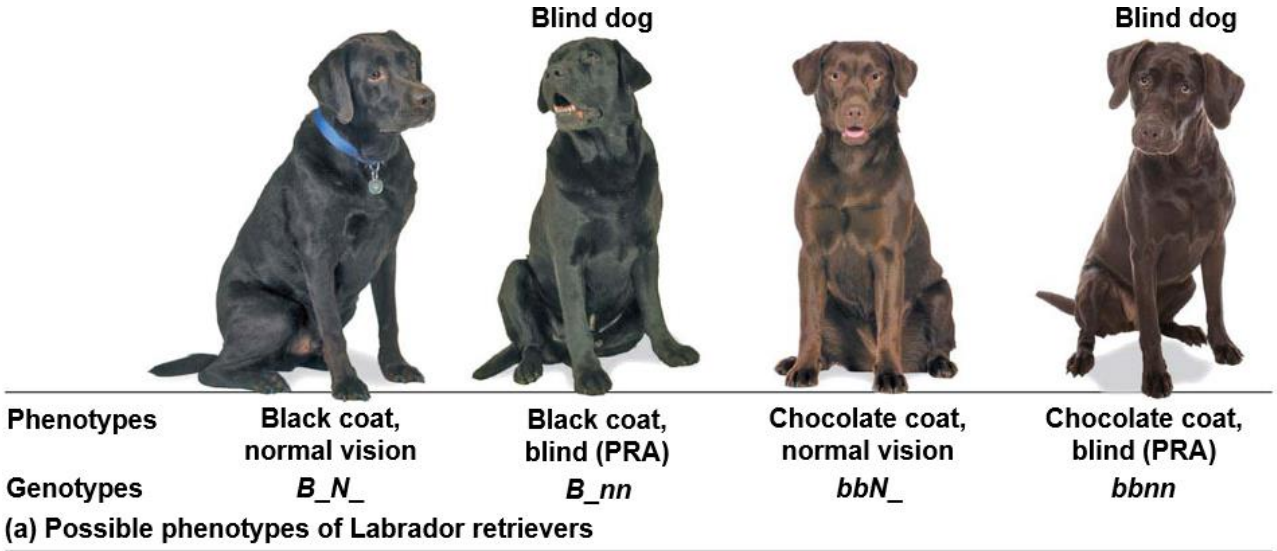


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Figure 9.9 Independent Assortment of Genes in Labrador Retrievers (2 of 3)

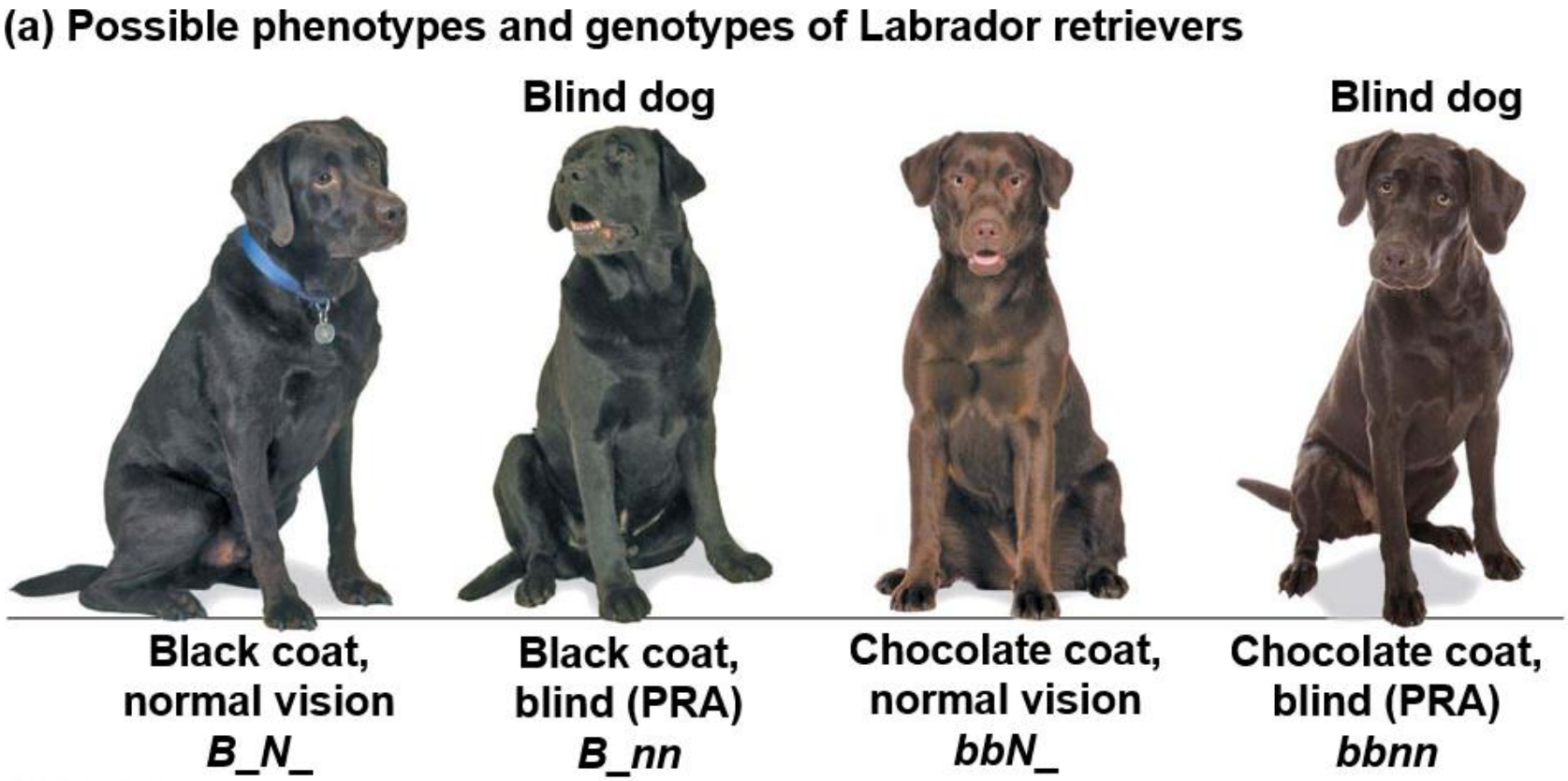
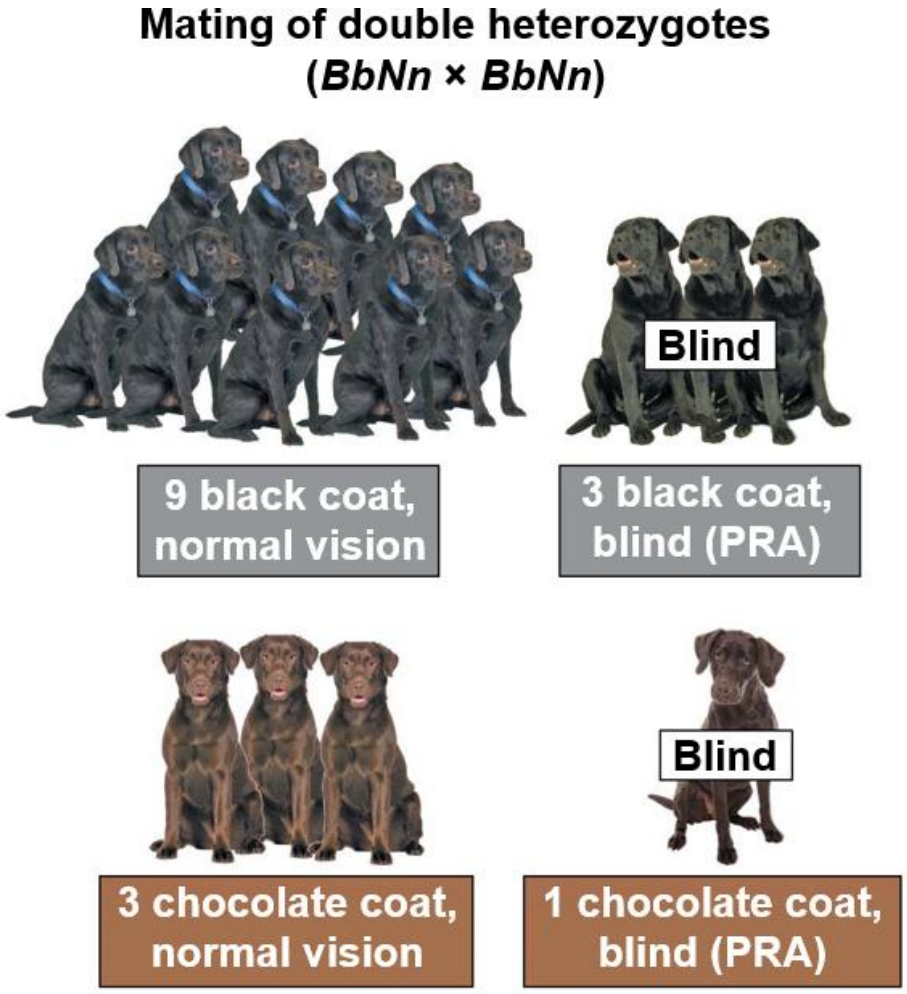


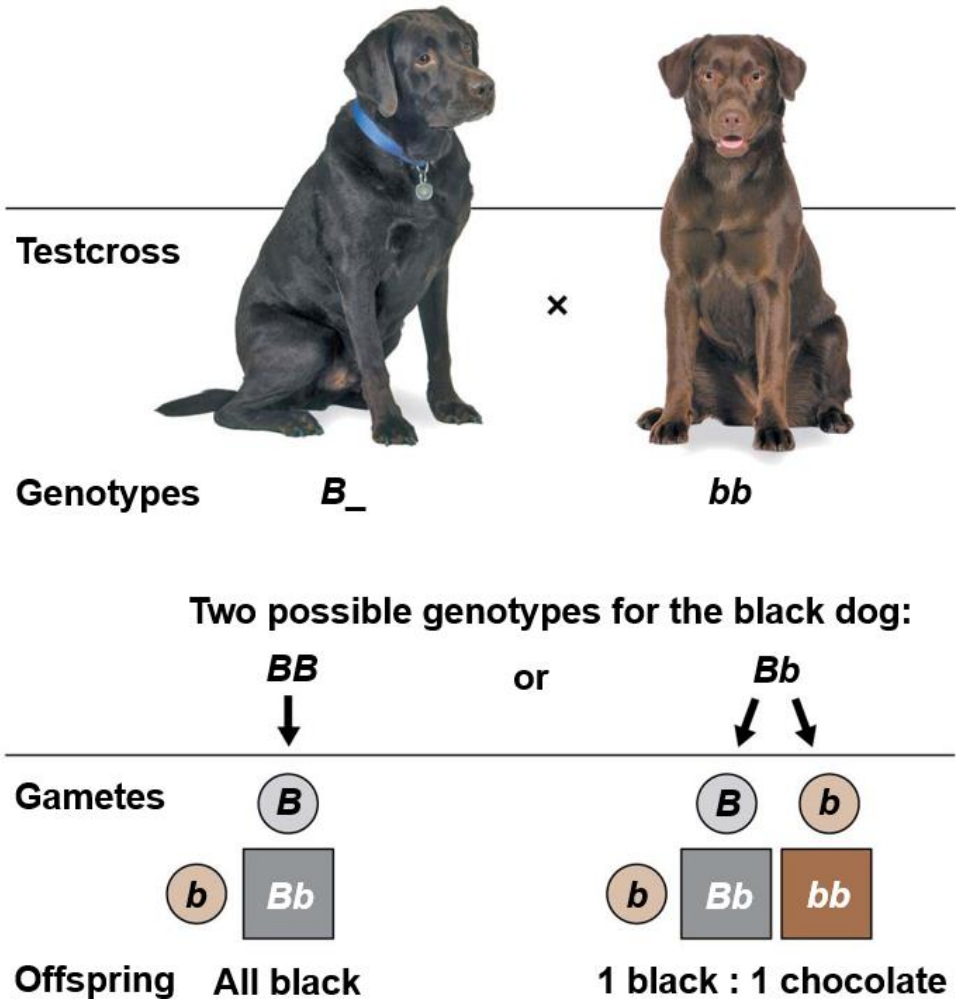
Figure 9.9 Independent Assortment of Genes in Labrador Retrievers (3 of 3)



Using a Testcross to Determine an Unknown Genotype

- A **testcross** is a mating between an individual of dominant phenotype (but unknown genotype) and a homozygous recessive individual.
- **Figure 9.10** shows the offspring that could result from such a mating.

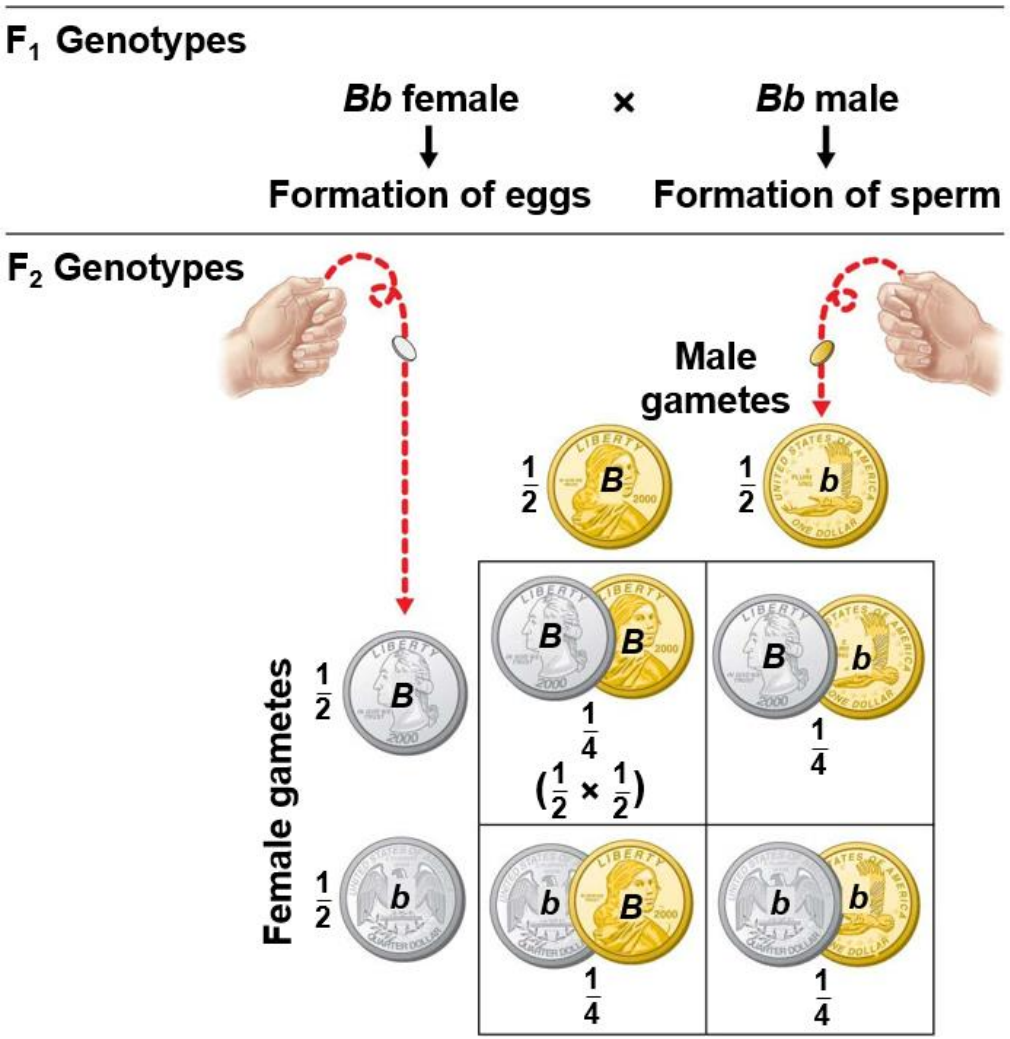
Figure 9.10 A Labrador Retriever Testcross



The Rules of Probability

- Mendel's strong background in mathematics helped him understand patterns of inheritance.
  - For instance, he understood that genetic crosses obey the rules of probability—the same rules that apply when tossing coins, rolling dice, or drawing cards.
  - 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 Segregation of Alleles and Fertilization as Chance Events



Family Pedigrees (1 of 3)

- Mendel's principles apply to the inheritance of many human traits.
- **Figure 9.12** illustrates alternative forms of three human characters that are each thought to be determined by simple dominant-recessive inheritance of one gene.

Figure 9.12 Examples of Inherited Human Traits Thought to Be Controlled by a Single Gene (1 of 4)

Figure 9.12 Examples of Inherited Human Traits Thought to Be Controlled by a Single Gene (2 of 4)

Freckles

No freckles

Figure 9.12 Examples of Inherited Human Traits Thought to Be Controlled by a Single Gene (3 of 4)

Widow's peak

Straight hairline

Figure 9.12 Examples of Inherited Human Traits Thought to Be Controlled by a Single Gene (4 of 4)

Free earlobe

Attached earlobe

Family Pedigrees (2 of 3)

- A trait that is dominant does not imply that it is either normal or more common than a recessive phenotype.
- **Wild-type traits** (those seen most often in nature) are not necessarily specified by dominant alleles.

Family Pedigrees (3 of 3)

- How can human genetics be studied?
- Geneticists
  - analyze the results of matings that have already occurred and
  - assemble this information into a family tree, called a **pedigree**.
- **Figure 9.13** shows a pedigree tracing the incidence of free versus attached earlobes.
- Mendel's laws enable us to deduce the genotypes for most of the people in the pedigree.

Figure 9.13 A Family Pedigree Showing Inheritance of Free Versus Attached Earlobes (1 of 4)

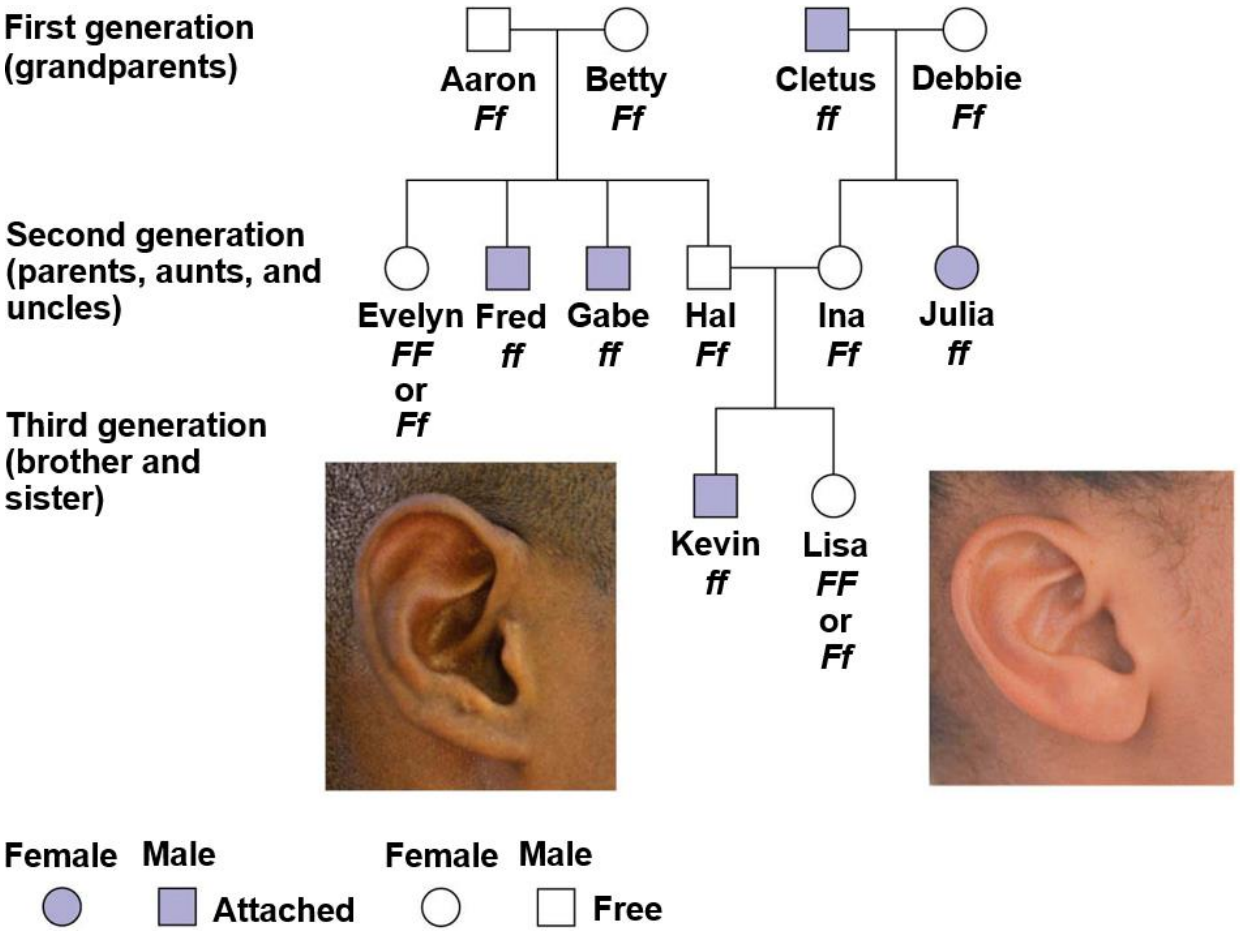


Figure 9.13 A Family Pedigree Showing Inheritance of Free Versus Attached Earlobes (2 of 4)

Figure 9.13 A Family Pedigree Showing Inheritance of Free Versus Attached Earlobes (3 of 4)

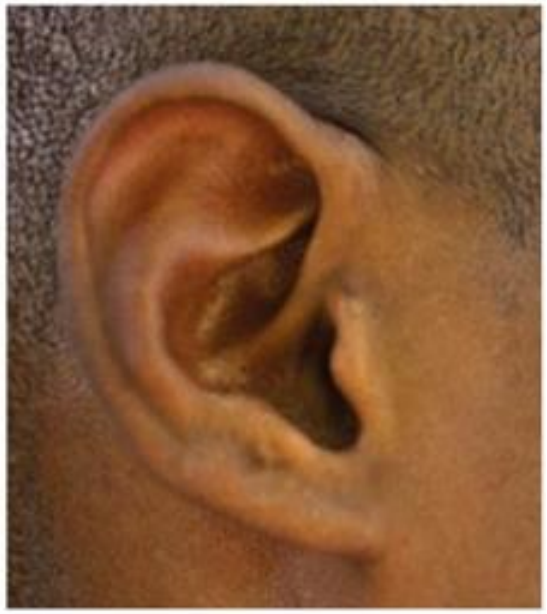
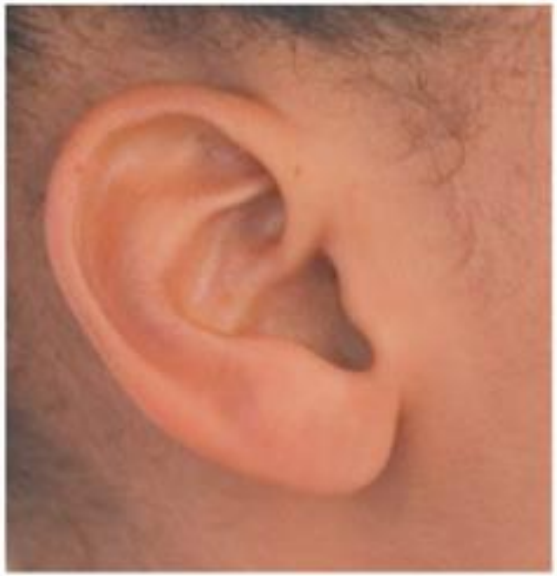


Figure 9.13 A Family Pedigree Showing Inheritance of Free Versus Attached Earlobes (4 of 4)



Human Disorders Controlled by a Single Gene

- Some human genetic disorders are known to be inherited as dominant or recessive traits controlled by a single gene, all located on autosomes, chromosomes other than the sex chromosomes X and Y.
- These disorders are listed in Table 9.1.

Table 9.1 Some Autosomal Disorders in People

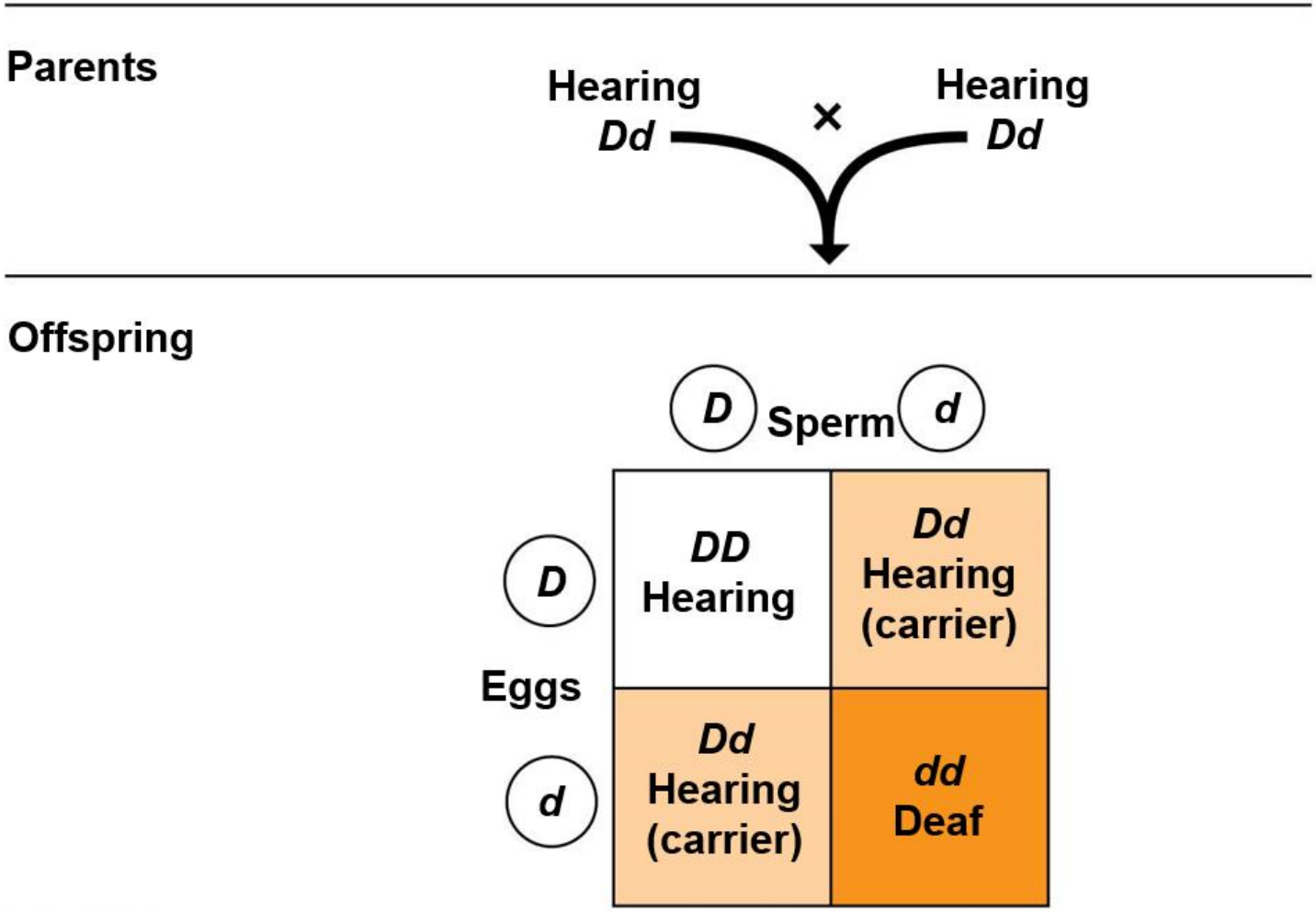
Table 9.1 Some Autosomal Disorders in People

Disorder	Major Symptoms
Recessive Disorders	
Albinism	Lack of pigment in skin, hair, and eyes
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation unless treated
Sickle-cell disease	Sickled red blood cells; damage to many tissues
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood
Dominant Disorders	
Achondroplasia	Dwarfism
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age
Hypercholesterolemia	Excess cholesterol in blood; heart disease

Recessive Disorders (1 of 2)

- Most human genetic disorders are recessive.
- Individuals who have the recessive allele but appear normal are **carriers** of the disorder.
- Using Mendel's laws, we can predict the fraction of affected offspring that is likely to result from a marriage between two carriers.

Figure 9.14 Predicted Offspring When Both Parents are Carriers for a Recessive Disorder



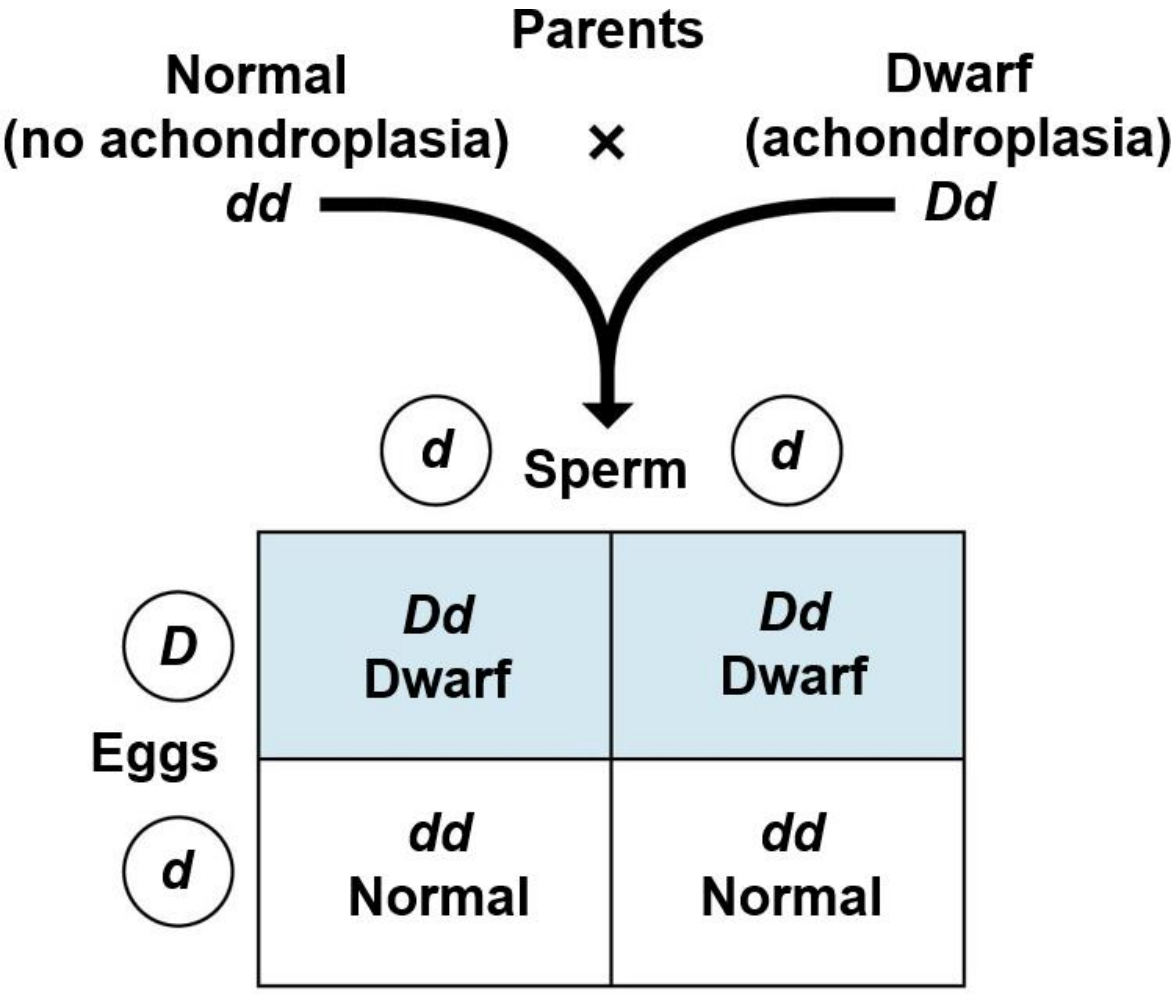
Recessive Disorders (2 of 2)

- **Cystic fibrosis** is
  - the most common lethal genetic disease in the United States and
  - caused by a recessive allele carried by about 1 in 31 Americans.

Dominant Disorders (1 of 2)

- A number of human disorders are caused by dominant alleles.
  - Achondroplasia is a form of dwarfism.
    - The homozygous dominant genotype (**AA**) causes death of the embryo.
    - Therefore, only heterozygotes (**Aa**), individuals with a single copy of the defective allele, have this disorder.
    - This also means that a person with achondroplasia has a 50% chance of passing the condition on to any children.

Figure 9.15 A Punnett Square Illustrating a Family with and Without Achondroplasia



Dominant Disorders (2 of 2)

- Dominant alleles that cause lethal disorders are much less common than lethal recessive alleles.
- One example is the allele that causes Huntington's disease, a degeneration of the nervous system that usually does not begin until middle age.
  - Once the deterioration of the nervous system begins, it is irreversible and inevitably fatal.
  - Because the allele for Huntington's disease is dominant, any child born to a parent with the allele has a 50% chance of inheriting the allele and the disorder.

The Process of Science: What Is the Genetic Basis of Coat Variation in Dogs? (1 of 2)

- **Observation**: Dogs come in a wide variety of physical types.
- **Question**: What is the genetic basis for canine coats?
- **Hypothesis**: A comparison of genes of a wide variety of dogs with different coats would identify the genes responsible.

Figure 9.16 Smooth Versus Wired Fox Terrier

The Process of Science: What Is the Genetic Basis of Coat Variation in Dogs? (2 of 2)

- **Prediction**: Mutations in just a few genes account for the coat appearance.
- **Experiment**: Researchers compared DNA sequences of 622 dogs from dozens of breeds.
- **Results**: Three genes in different combinations produced seven different coat appearances, from very short hair to full, thick, wired hair.

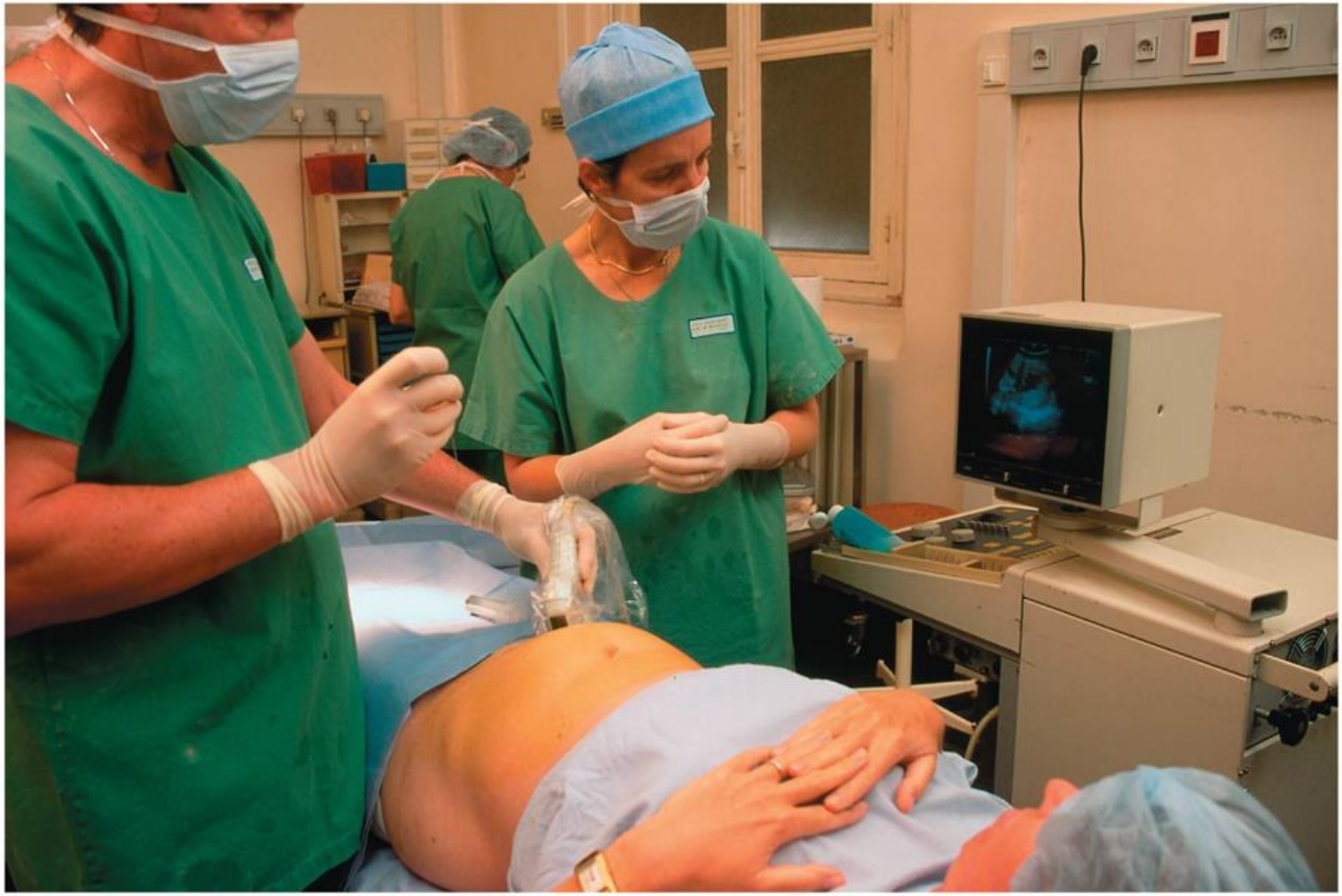
Genetic Testing (1 of 3)

- Today many tests can detect the presence of disease-causing alleles.
- Most genetic tests are performed during pregnancy if the prospective parents are aware that they have an increased risk of having a baby with a genetic disease.
  - In amniocentesis, a physician uses a needle to extract about 2 teaspoons of the fluid that bathes the developing fetus.

Genetic Testing (2 of 3)

- In chorionic villus sampling, a physician inserts a narrow, flexible tube through the mother's vagina and into her uterus, removing some placental tissue.
- Newer genetic screening procedures involve isolating tiny amounts of fetal cells or DNA released into the mother's bloodstream. These newer technologies are gradually replacing more invasive screening methods because they
  - are more accurate and
  - can be performed earlier than other tests.

Figure 9.17 Amniocentesis



Genetic Testing (3 of 3)

- Once cells are obtained, they can be screened for genetic diseases.
- Patients seeking genetic testing should receive counseling both before and after to explain the test and to help them cope with the results.

Variations on Mendel's Laws

- Mendel's two laws explain inheritance in terms of genes that are passed along from generation to generation according to simple rules of probability.
  - These laws are valid for all sexually reproducing organisms.
  - But Mendel's laws stop short of explaining some patterns of genetic inheritance.
  - In fact, for most sexually reproducing organisms, cases in which Mendel's rules can strictly account for the patterns of inheritance are relatively rare.
  - More often, the observed inheritance patterns are more complex.

Incomplete Dominance in Plants and People (1 of 3)

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

Figure 9.18 Incomplete Dominance in Snapdragons (1 of 3)

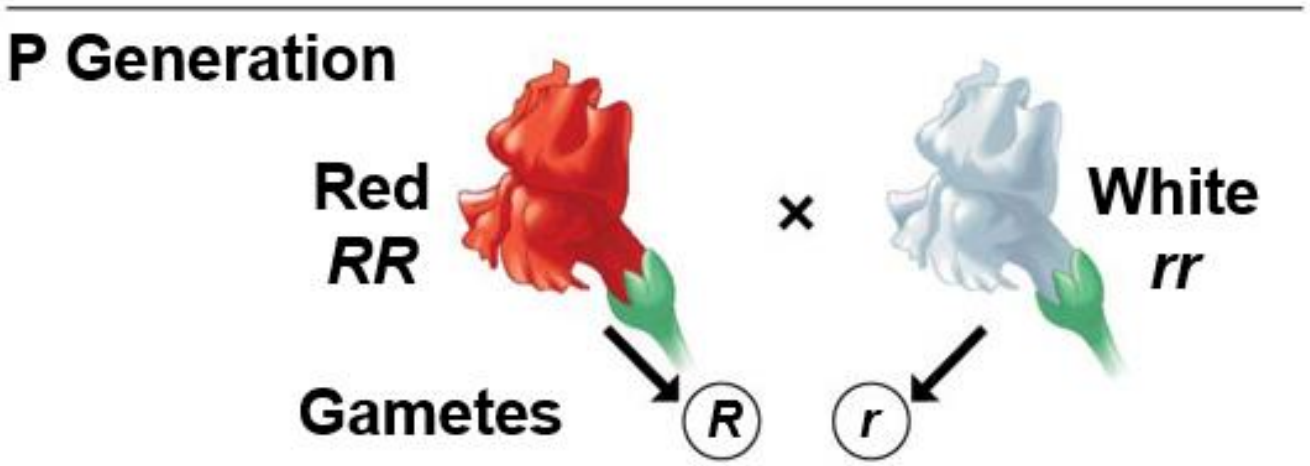


Figure 9.18 Incomplete Dominance in Snapdragons (2 of 3)

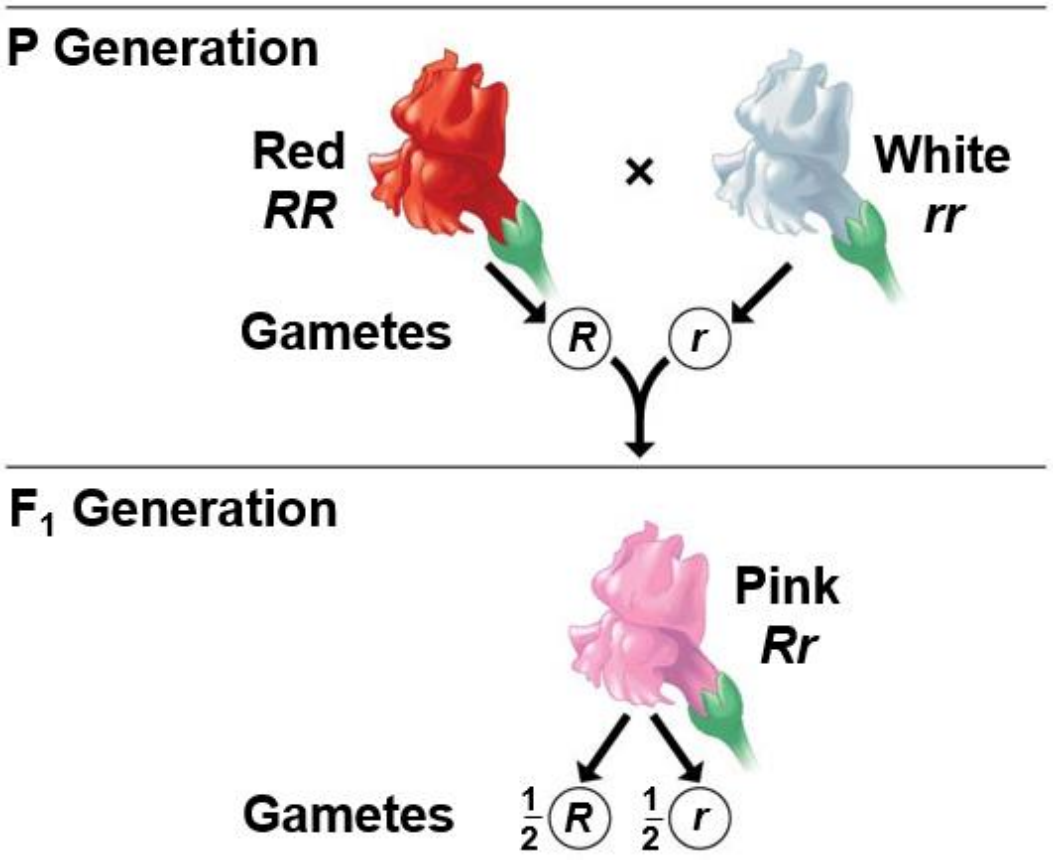
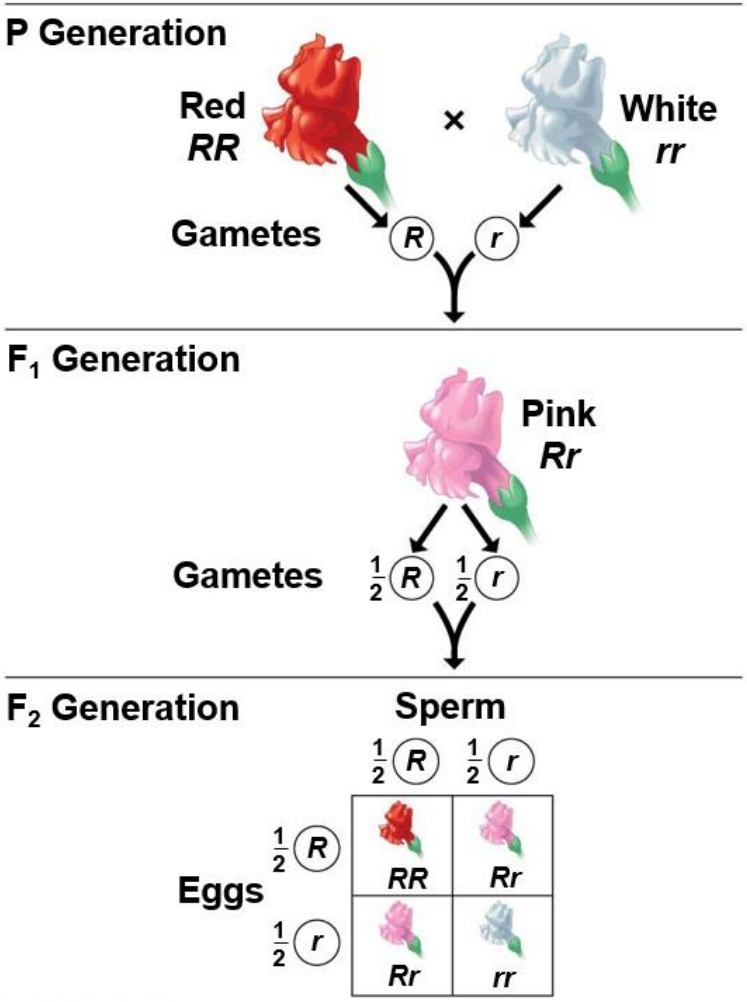


Figure 9.18 Incomplete Dominance in Snapdragons (3 of 3)



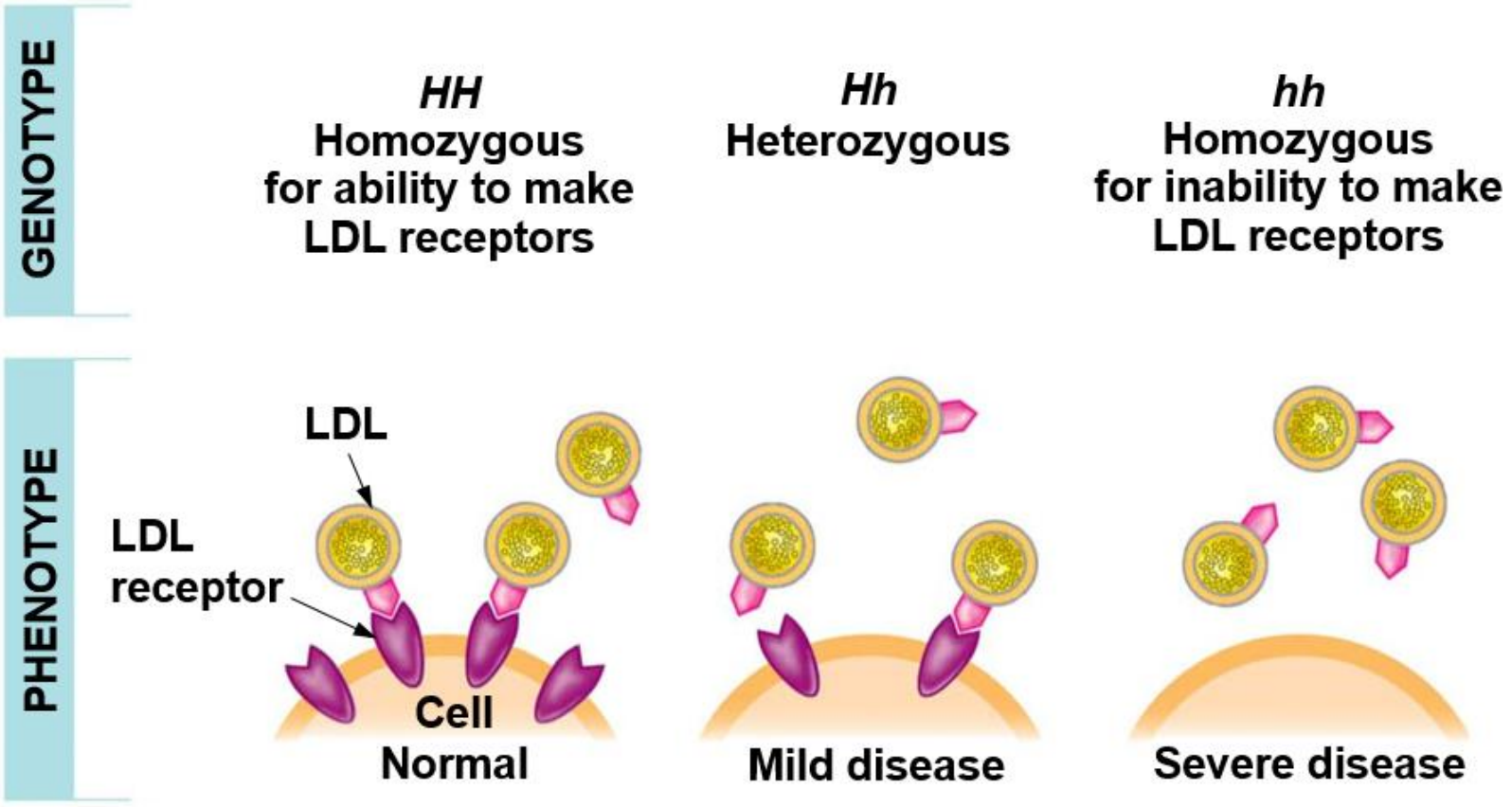
Incomplete Dominance in Plants and People (2 of 3)

- Hypercholesterolemia
  - is a human trait that is an example of incomplete dominance and
  - is characterized by dangerously high levels of cholesterol in the blood.

Incomplete Dominance in Plants and People (3 of 3)

- In hypercholesterolemia,
  - heterozygotes have blood cholesterol levels about twice normal, while
  - homozygotes have about five times the normal amount of blood cholesterol and may have heart attacks as early as age 2.

Figure 9.19 Incomplete Dominance in Human Hypercholesterolemia



ABO Blood Groups: An Example of Multiple Alleles and Codominance (1 of 4)

- The **ABO blood groups** in humans involve three alleles of a single gene.
  - Various combinations of these three alleles produce four phenotypes: A person's blood type may be A, B, AB, or O.
  - These letters refer to two carbohydrates, designated A and B, that may be found on the surface of red blood cells.

Figure 9.20 Multiple Alleles for the ABO Blood Groups (1 of 4)

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	$I^A I^A$ or $I^A i$	Carbohydrate A	Anti-B				
B	$I^B I^B$ or $I^B i$	Carbohydrate B	Anti-A				
AB	$I^A I^B$		—				
O	ii		Anti-A Anti-B				

Figure 9.20 Multiple Alleles for the ABO Blood Groups (2 of 4)

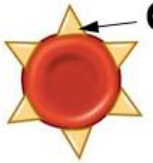
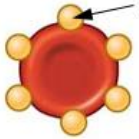
Blood Group (Phenotype)	Genotypes	Red Blood Cells	Antibodies Present in Blood
A	$I^A I^A$ or $I^A i$	 Carbohydrate A	Anti-B
B	$I^B I^B$ or $I^B i$	 Carbohydrate B	Anti-A
AB	$I^A I^B$		—
O	ii		Anti-A Anti-B

Figure 9.20 Multiple Alleles for the ABO Blood Groups (3 of 4)

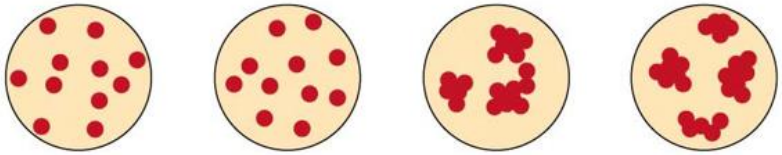
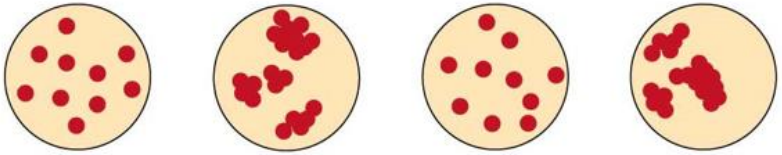
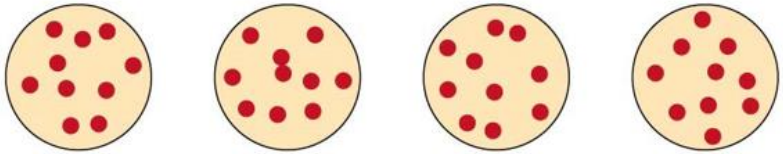
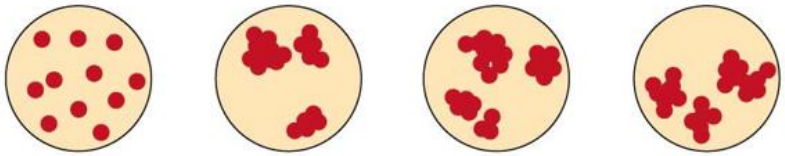
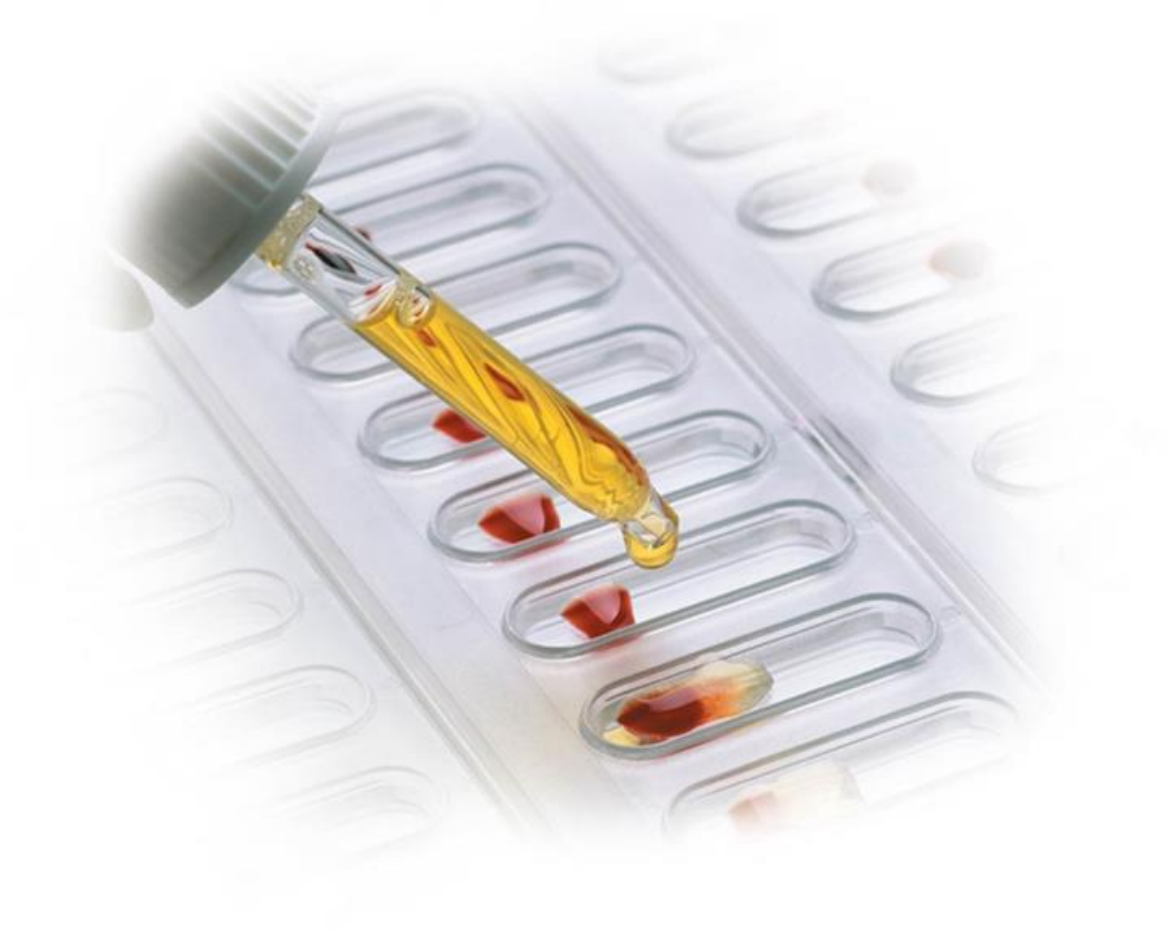
Blood Group (Phenotype)	Reactions When Blood from Groups Below Is Mixed with Antibodies from Groups at Left			
	O	A	B	AB
A				
B				
AB				
O				

Figure 9.20 Multiple Alleles for the ABO Blood Groups (4 of 4)



ABO Blood Groups: An Example of Multiple Alleles and Codominance (2 of 4)

- Matching compatible blood groups is critical for safe blood transfusions. If a donor's blood cells have a carbohydrate (A or B) that is foreign to the recipient, then the recipient's immune system produces blood proteins called antibodies that bind to the foreign carbohydrates and cause the donor blood cells to clump together, potentially killing the recipient.

ABO Blood Groups: An Example of Multiple Alleles and Codominance (3 of 4)

- The four blood groups result from various combinations of the three different alleles:
  - I^A (for the ability to make substance A),
  - I^B (for B), and
  - i (for neither A nor B).
- Each person inherits one of these alleles from each parent.

ABO Blood Groups: An Example of Multiple Alleles and Codominance (4 of 4)

- Because there are three alleles, there are six possible genotypes, as listed in Figure 9.20.
- Both the I^A and I^B alleles are dominant to the *i* allele.
- People of genotype I^AI^B make both carbohydrates. In other words, the I^A and I^B alleles are **codominant**, meaning that both alleles are expressed in heterozygous individuals (I^AI^B) who have type AB blood.

Structure/Function: Pleiotropy and Sickle-Cell Disease

- **Pleiotropy** is when one gene influences several characters.
- Sickle-cell disease
  - exhibits pleiotropy,
  - results in abnormal hemoglobin proteins, and
  - causes disk-shaped red blood cells to deform into a sickle shape with jagged edges.
- In most cases, only people who are homozygous for the sickle-cell allele have sickle-cell disease.

Figure 9.21 Sickle-Cell Disease: Multiple Effects of a Single Human Gene (1 of 2)

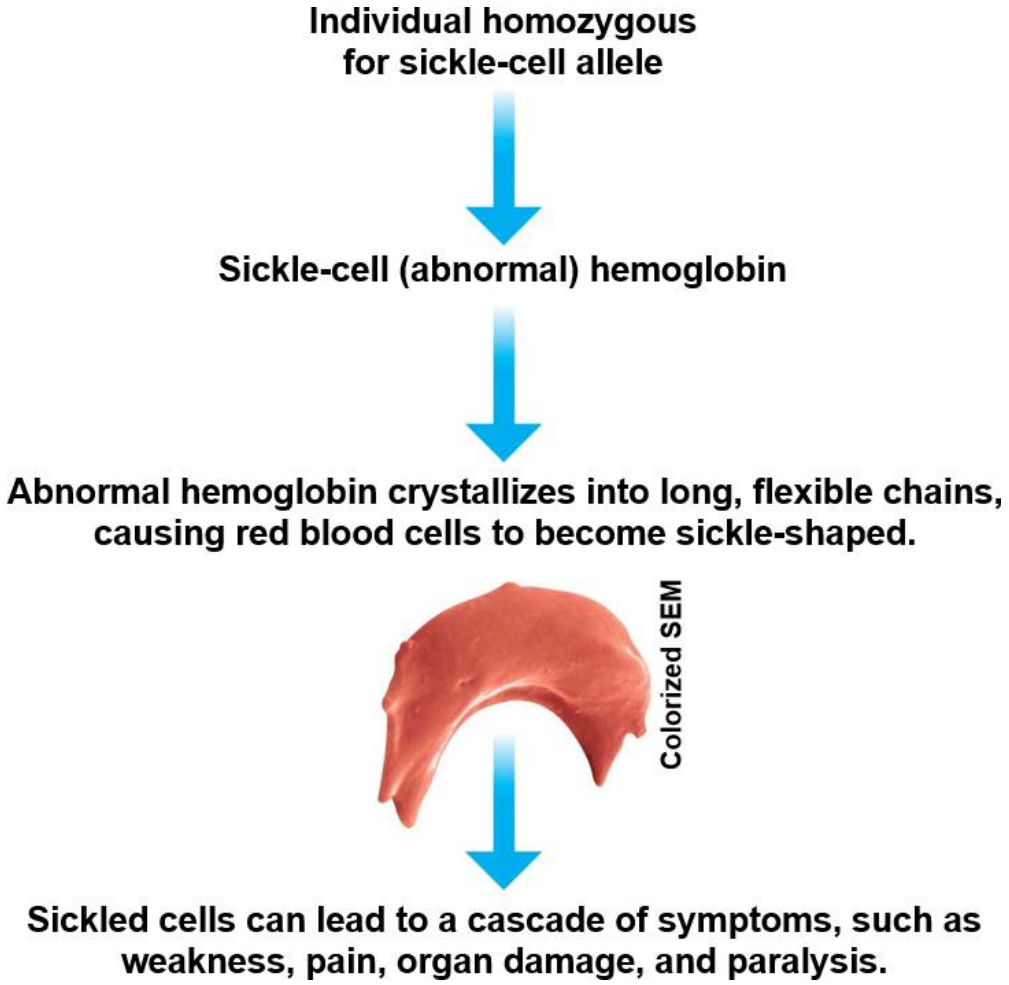
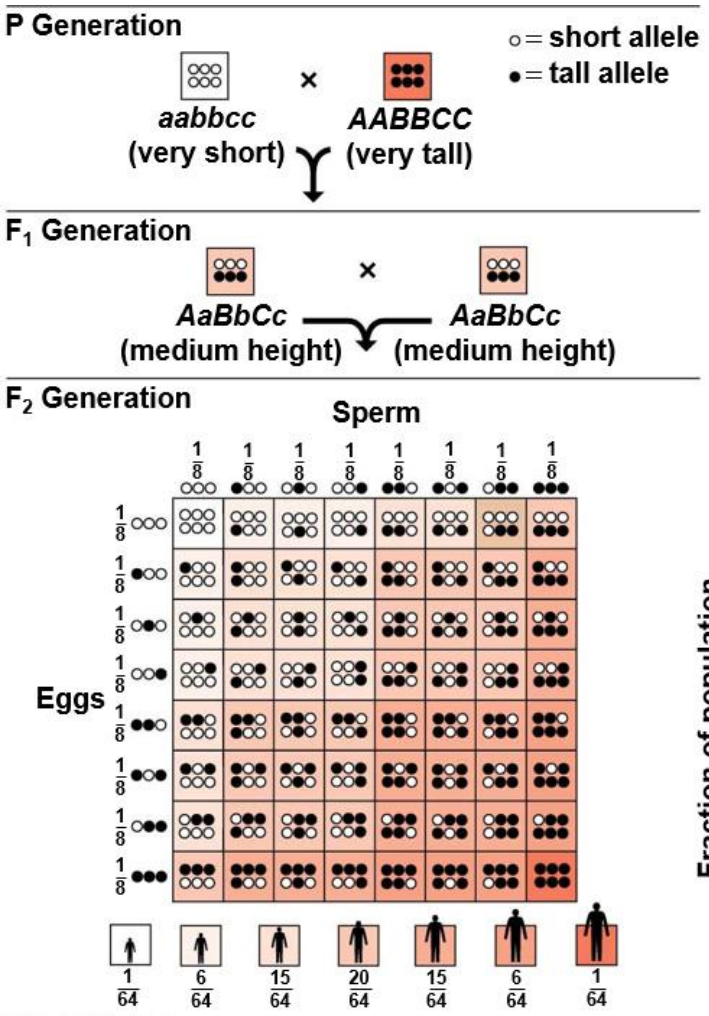


Figure 9.21 Sickle-Cell Disease: Multiple Effects of a Single Human Gene (2 of 2)

Polygenic Inheritance

- **Polygenic inheritance** is
  - the additive effects of two or more genes on a single phenotypic character and
  - the logical opposite of pleiotropy, in which one gene affects several characters.
- There is evidence that height in people is controlled by several genes that are inherited separately. (Actually, human height is probably affected by a great number of genes, but we'll simplify here.)

Figure 9.22 A Model for Polygenic Inheritance of Height (1 of 4)



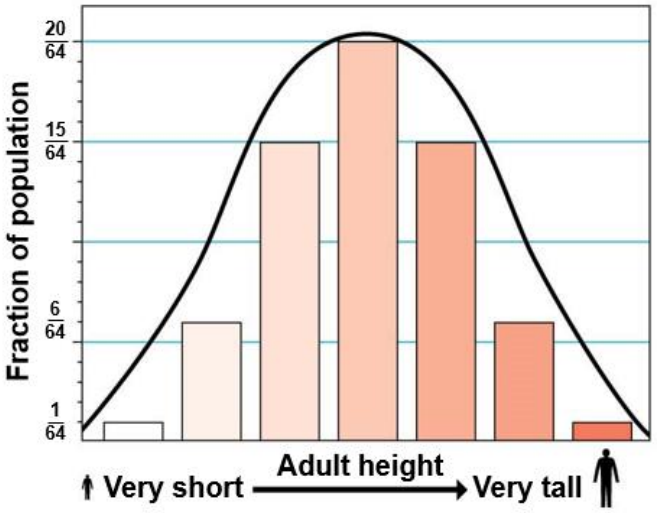


Figure 9.22 A Model for Polygenic Inheritance of Height (2 of 4)

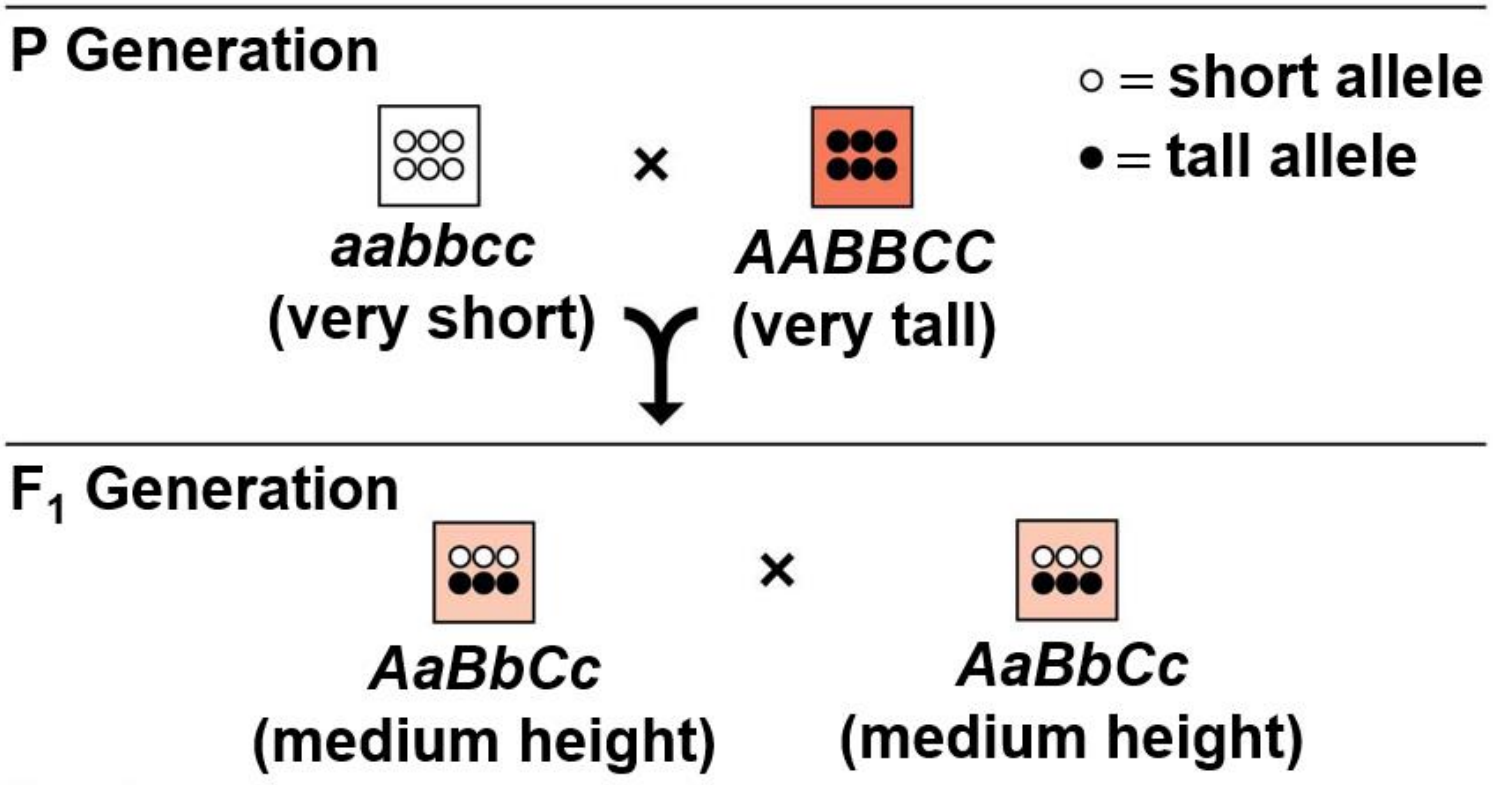


Figure 9.22 A Model for Polygenic Inheritance of Height (3 of 4)

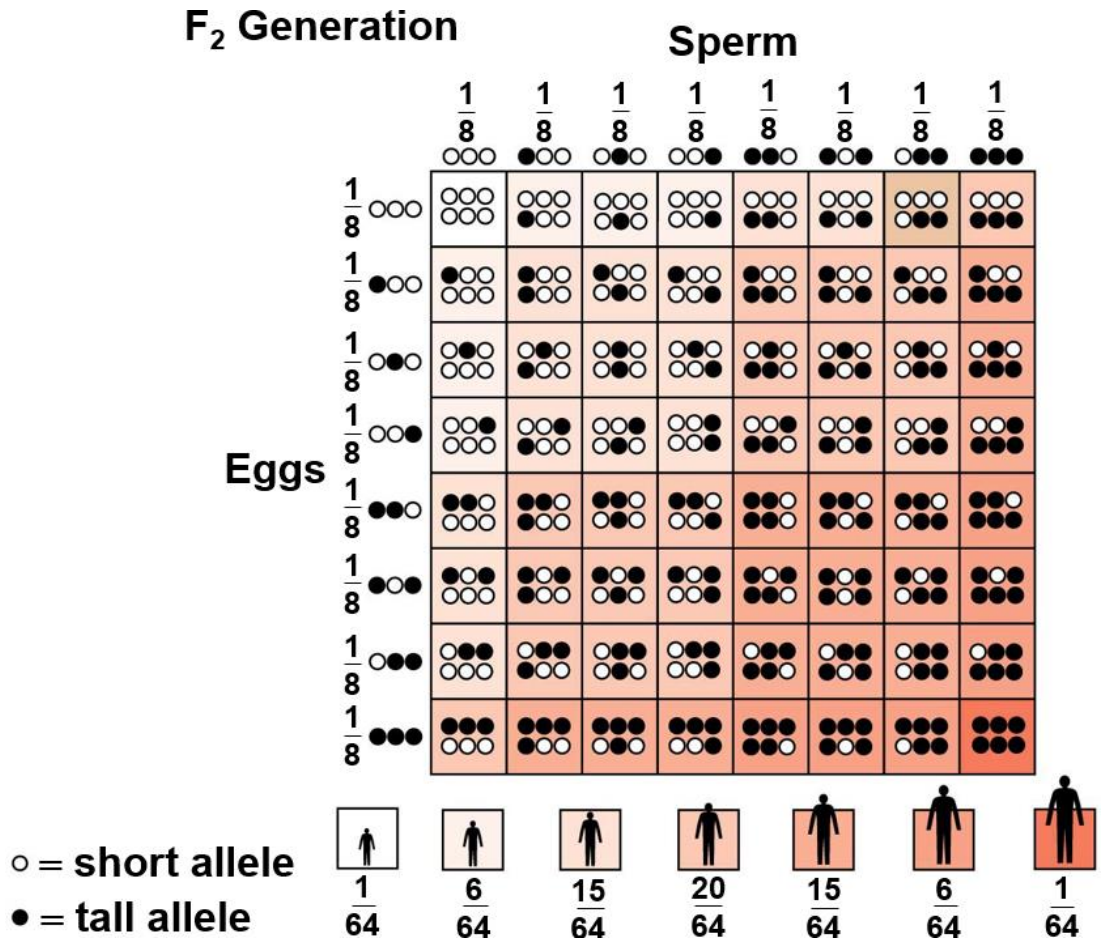
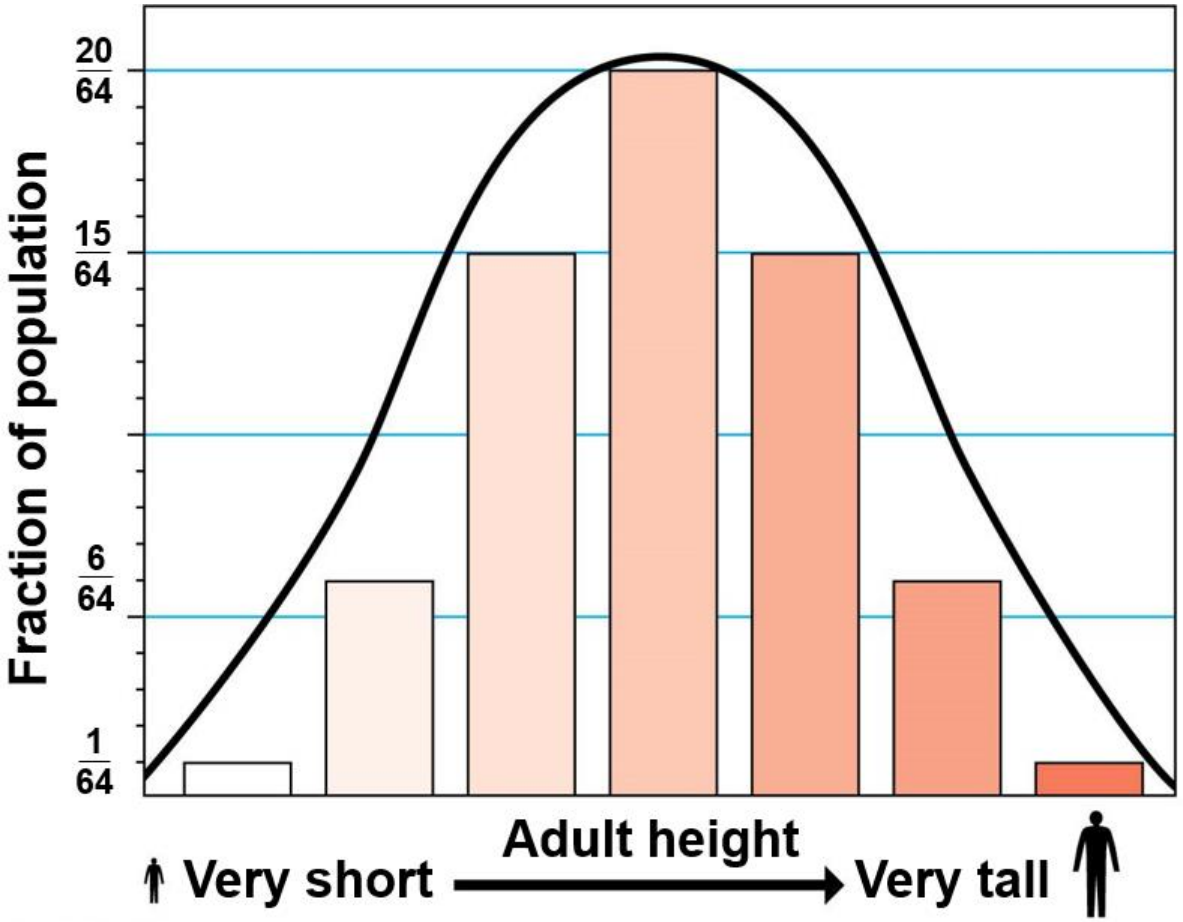


Figure 9.22 A Model for Polygenic Inheritance of Height (4 of 4)



Epigenetics and the Role of Environment (1 of 4)

- Many phenotypic characters result from a combination of heredity and environment.
  - Whether human characters are more influenced by genes or by the environment—nature or nurture—is a very old and hotly contested issue.
  - Spending time with identical twins will convince anyone that environment, and not just genes, affects a person's traits.

Figure 9.23 As a result of environmental influences, even identical twins can look different (1 of 2)

Figure 9.23 As a result of environmental influences, even identical twins can look different (2 of 2)

Epigenetics and the Role of Environment (2 of 4)

- In general, only genetic influences are inherited and effects of the environment are not passed to the next generation.
- In recent years, however, biologists have begun to recognize the importance of **epigenetic inheritance**, the transmission of traits by mechanisms not directly involving DNA sequence.

Epigenetics and the Role of Environment (3 of 4)

- For example, components of chromosomes can be chemically modified by adding or removing chemical groups on the DNA and/or protein components of chromosomes.
- Over a lifetime, the environment plays a role in these changes, which may explain how one identical twin can suffer from a genetically based disease whereas the other twin does not, despite their identical genomes.

Epigenetics and the Role of Environment (4 of 4)

- Epigenetic modifications—and the changes in gene activity that result—may even be carried on to the next generation.
- Unlike alterations to the DNA sequence, chemical changes to the chromosomes can be reversed.

The Chromosomal Basis of Inheritance (1 of 2)

- The **chromosome theory of inheritance** states that
  - genes are located at specific positions (loci) on chromosomes and
  - the behavior of chromosomes during meiosis and fertilization accounts for inheritance patterns.

The Chromosomal Basis of Inheritance (2 of 2)

- It is **chromosomes** that
  - undergo segregation and independent assortment during meiosis and
  - account for Mendel's laws.

Figure 9.24 The Chromosomal Basis of Mendel's Laws (1 of 3)

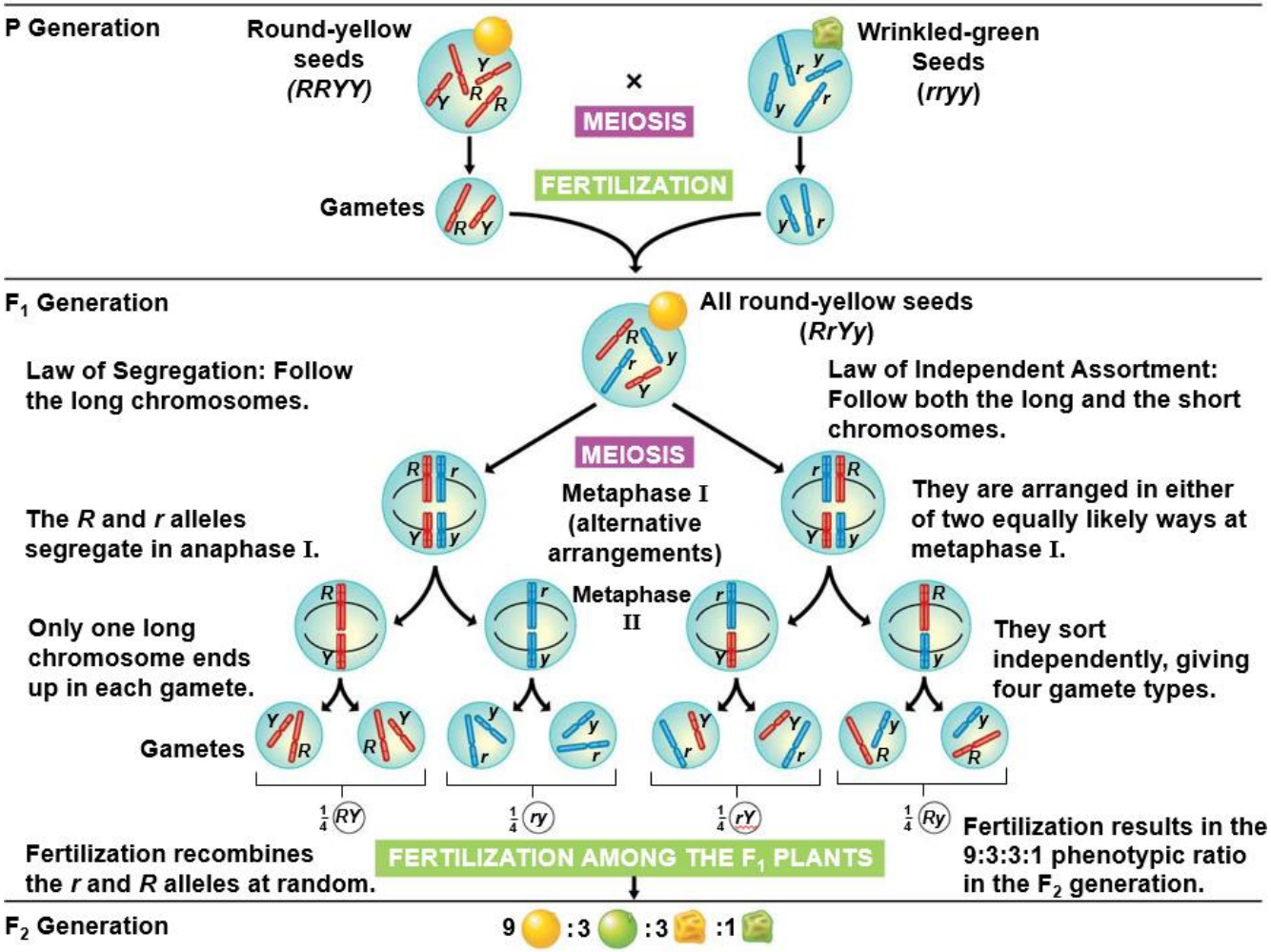


Figure 9.24 The Chromosomal Basis of Mendel's Laws (2 of 3)

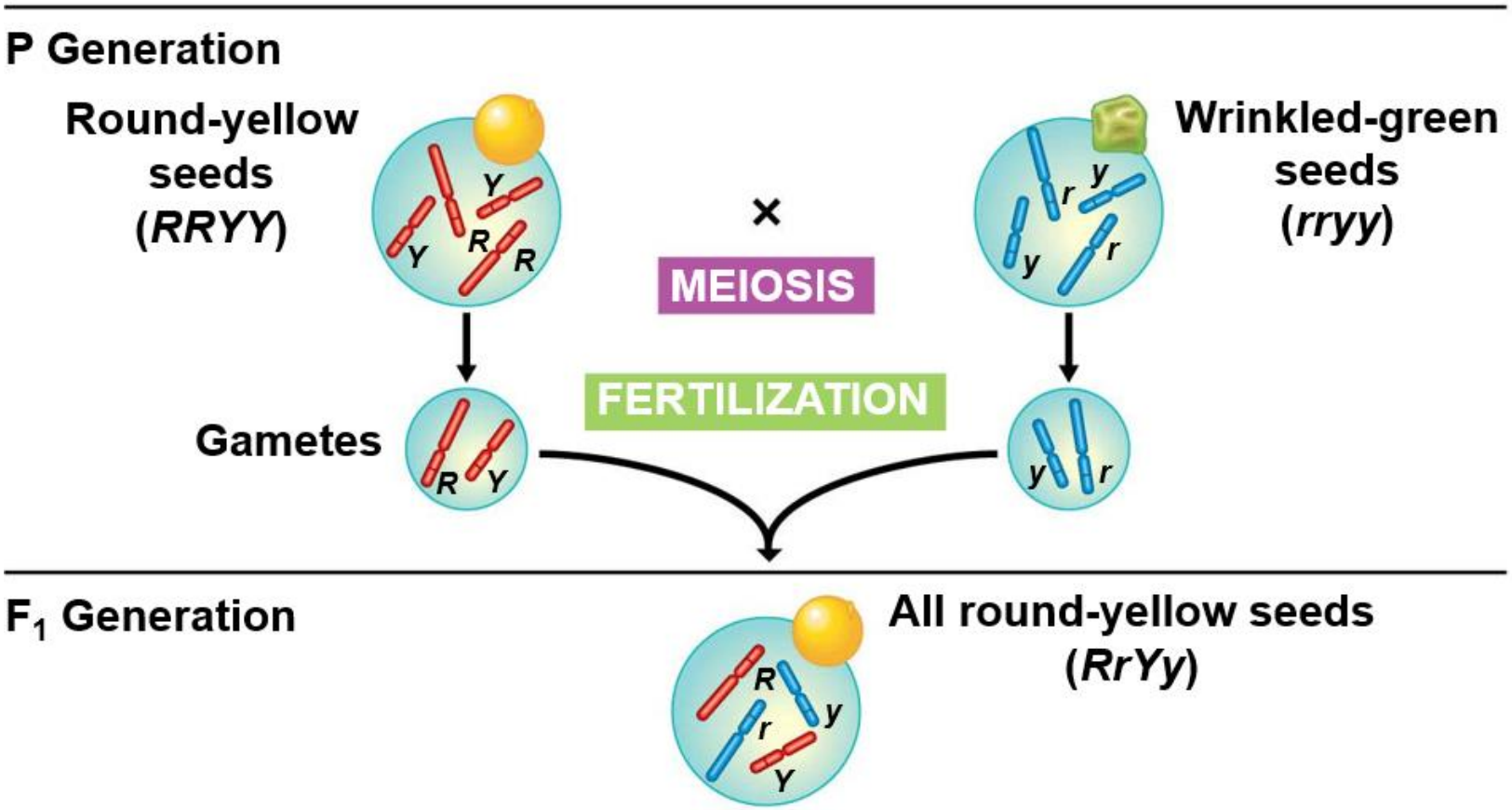
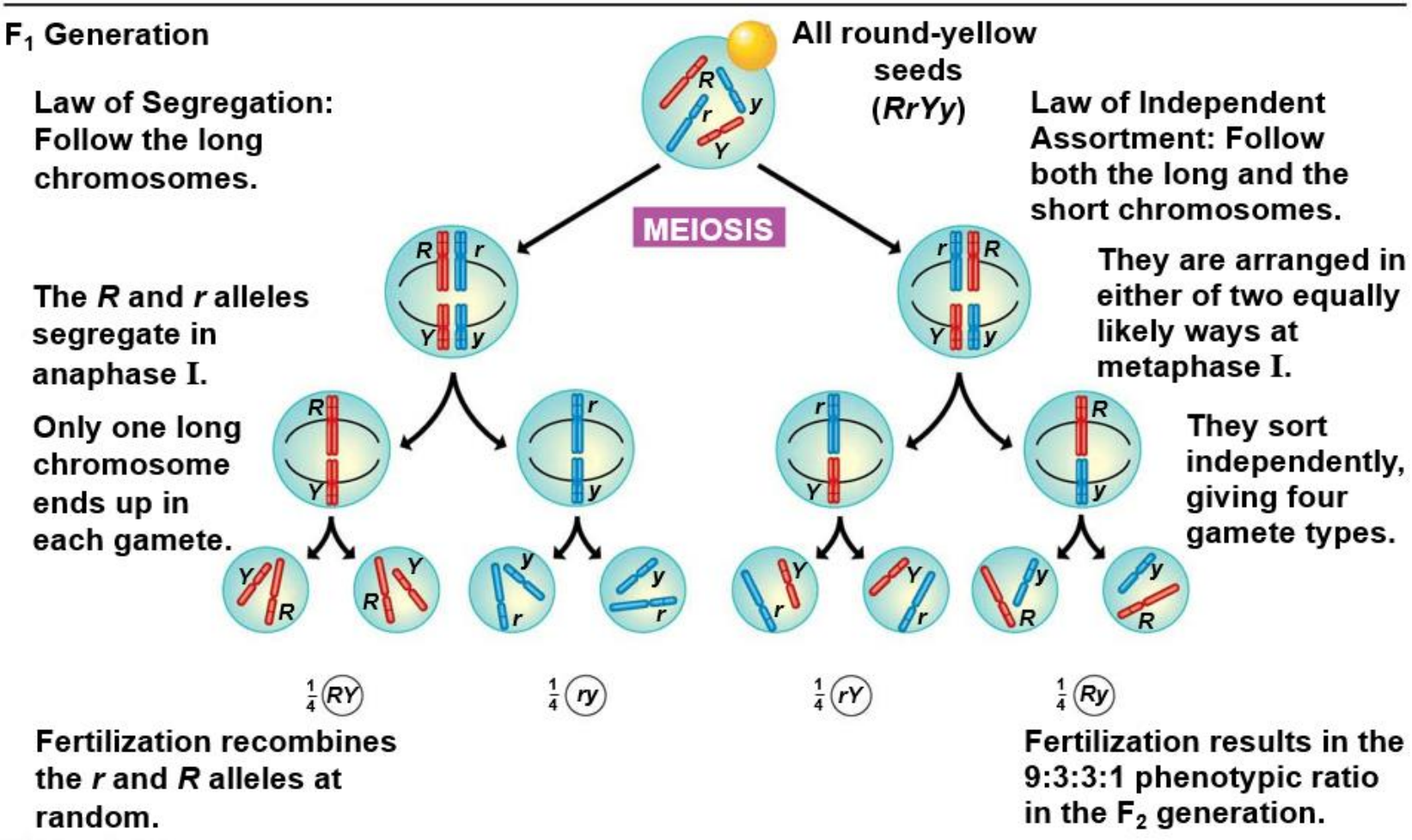


Figure 9.24 The Chromosomal Basis of Mendel's Laws (3 of 3)



Pearson

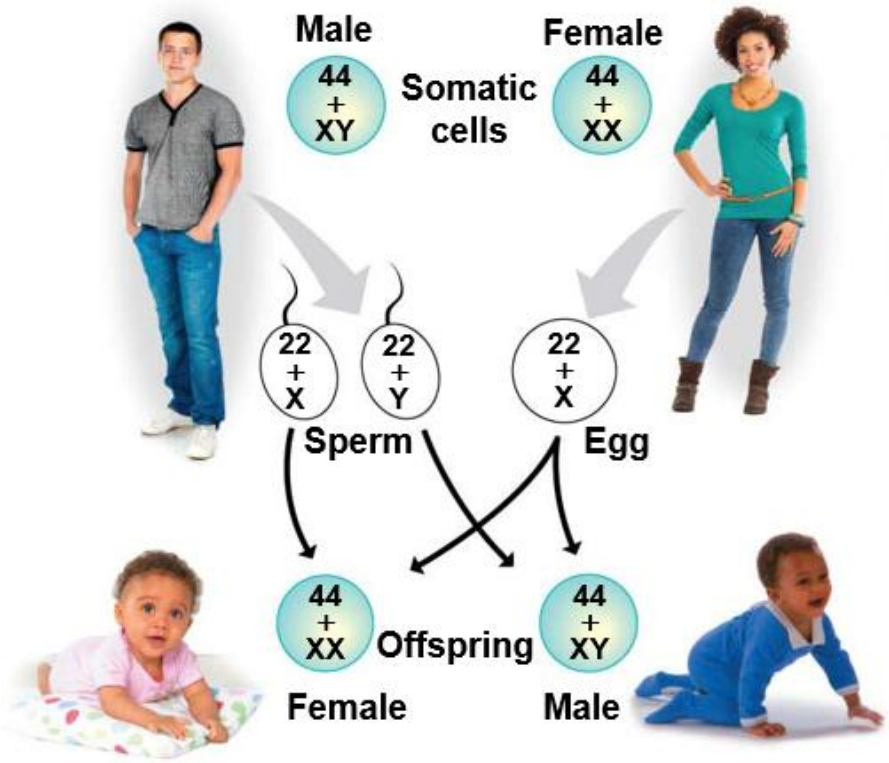
Linked Genes

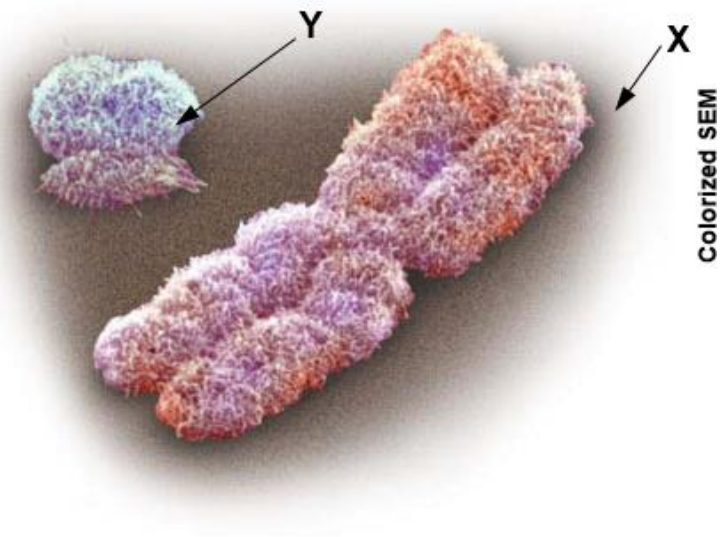
- **Linked genes**
  - are located near each other on the same chromosome and
  - tend to travel together during meiosis and fertilization.
- Such genes are often inherited as a set and therefore often do not follow Mendel's law of independent assortment.

Sex Determination in Humans

- Many animals, including all mammals, have a pair of sex chromosomes, designated X and Y, that determine an individual's sex.
  - Individuals with one X chromosome and one Y chromosome are males.
  - XX individuals are females.
- Human males and females both have 44 autosomes (chromosomes other than sex chromosomes).

Figure 9.25 The Chromosomal Basis of Sex Determination in Humans (1 of 3)





Pearson

Figure 9.25 The Chromosomal Basis of Sex Determination in Humans (2 of 3)

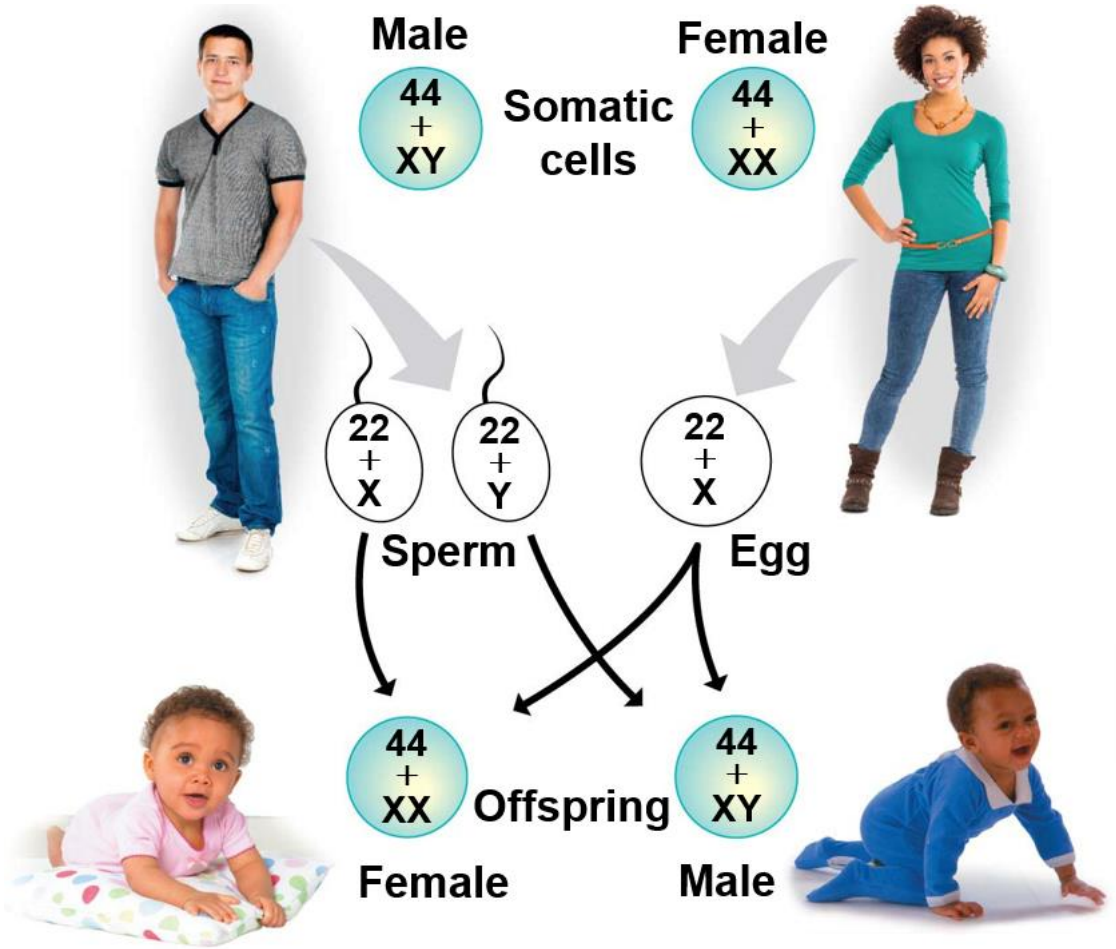
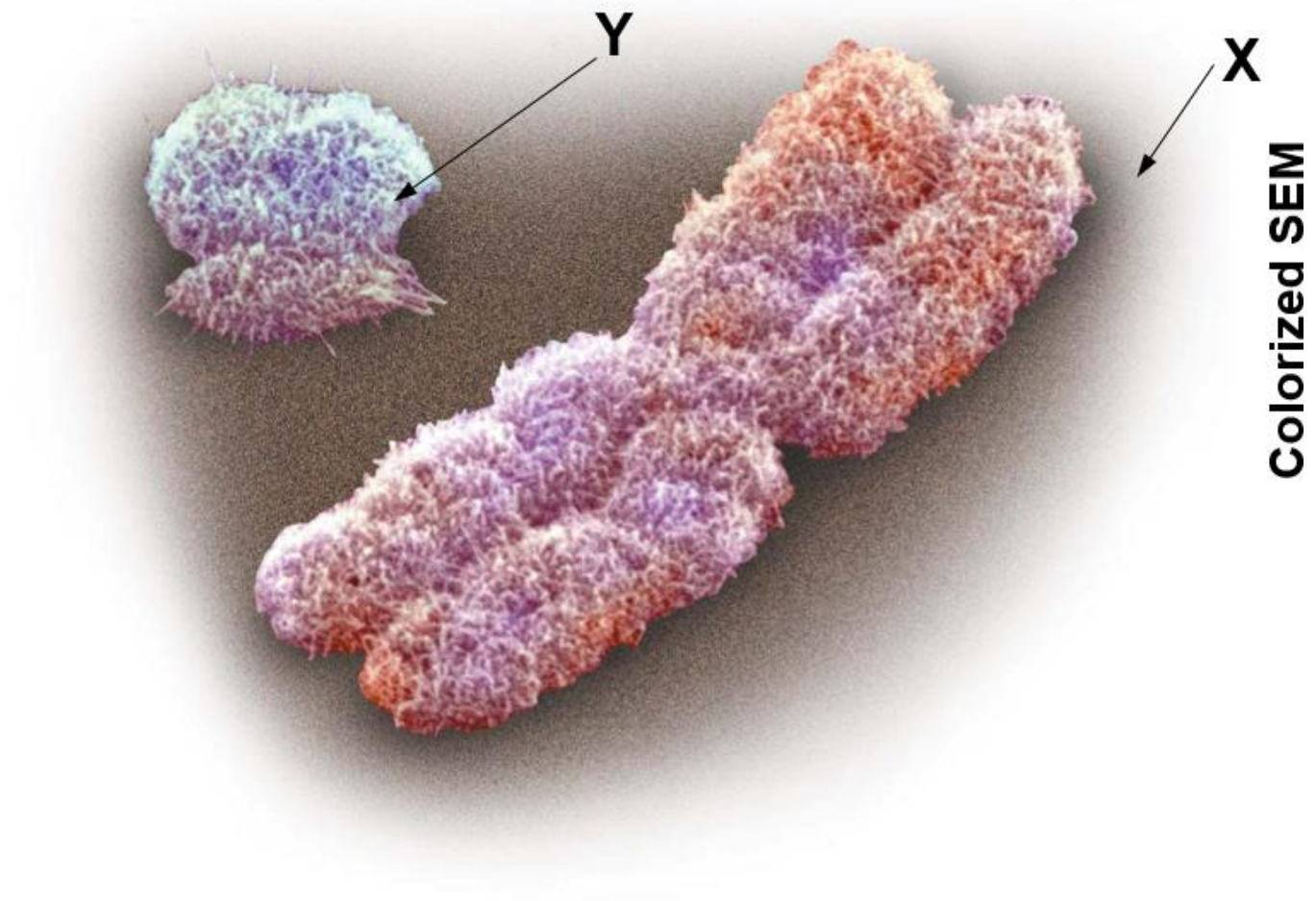


Figure 9.25 The Chromosomal Basis of Sex Determination in Humans (3 of 3)



Sex-Linked Genes (1 of 5)

- A gene located on a sex chromosome is called a **sex-linked gene**.
  - Most sex-linked genes are found on the X chromosome.

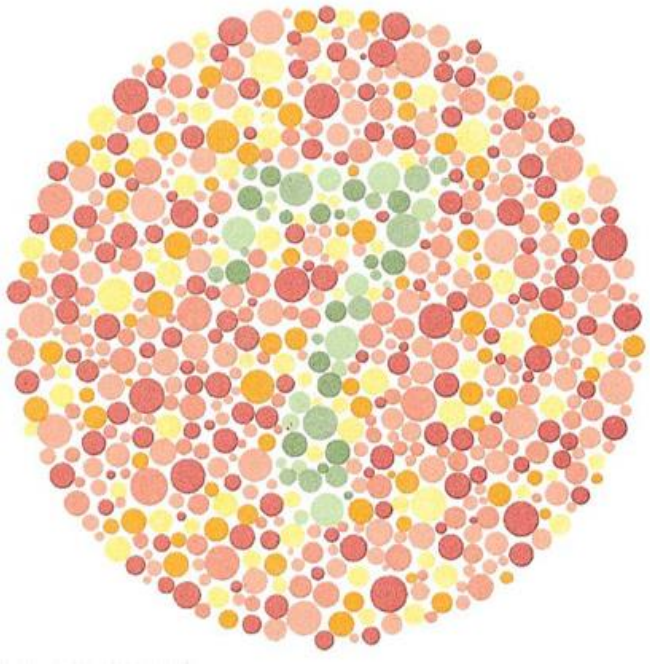
Sex-Linked Genes (2 of 5)

- A number of human conditions, including red-green colorblindness, hemophilia, and a type of muscular dystrophy, result from sex-linked recessive alleles.

Sex-Linked Genes (3 of 5)

- Red-green colorblindness is a common sex-linked disorder caused by a malfunction of light-sensitive cells in the eyes.
  - Mostly males are affected, but heterozygous females have some defects, too.
  - **Figure 9.26** shows a simple test for red-green colorblindness.

Figure 9.26 A Test for Red-Green Colorblindness



Sex-Linked Genes (4 of 5)

- Because they are located on the sex chromosomes, sex-linked genes exhibit unusual inheritance patterns.
  - **Figure 9.27a** illustrates what happens when a colorblind male has offspring with a homozygous female with normal color vision.
  - **Figure 9.27b** illustrates what happens if a female carrier mates with a male who has normal color vision.
  - Because the colorblindness allele is recessive, a female will be colorblind only if she receives that allele on both X chromosomes.

Figure 9.27 Inheritance of Colorblindness, a Sex-Linked Recessive Trait (1 of 4)

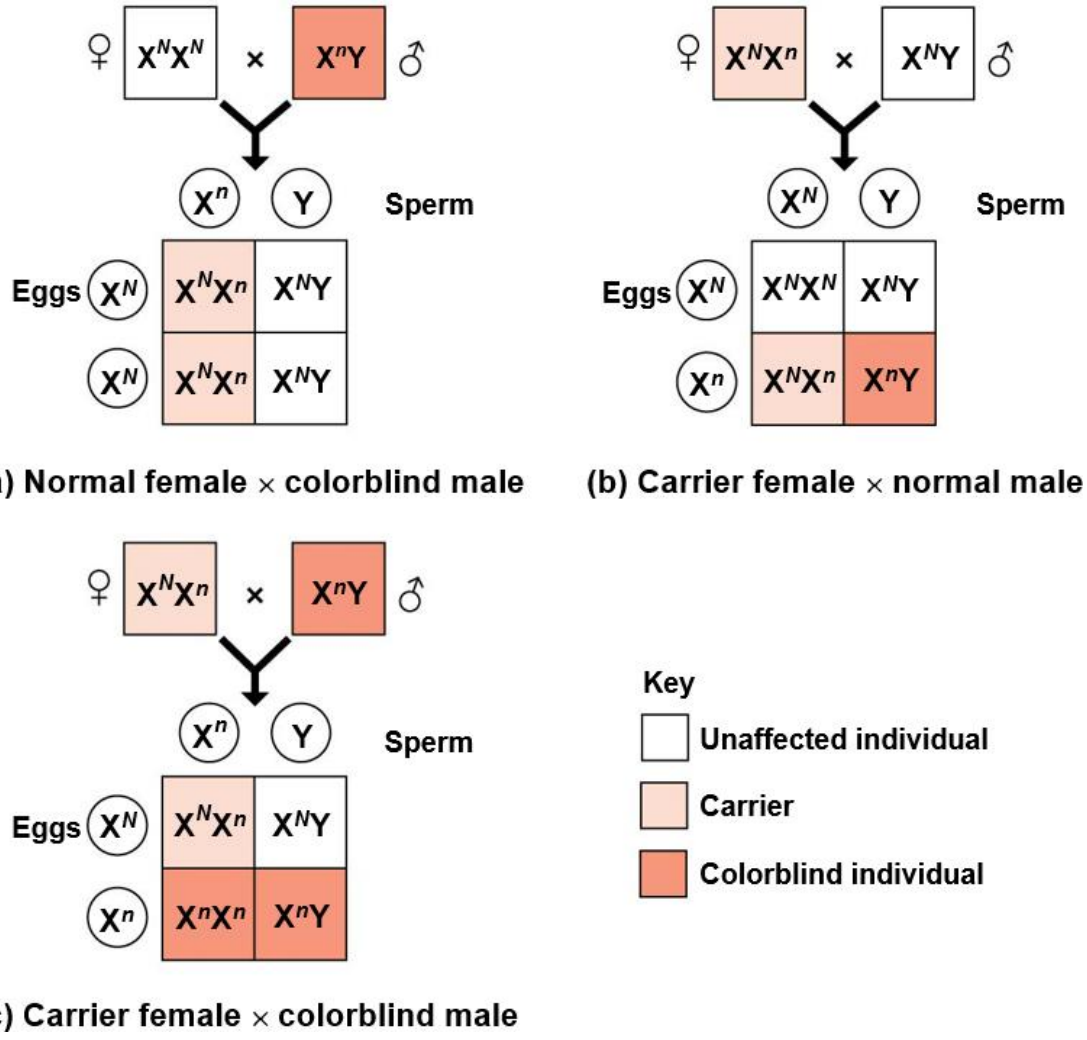


Figure 9.27 Inheritance of Colorblindness, a Sex-Linked Recessive Trait (2 of 4)

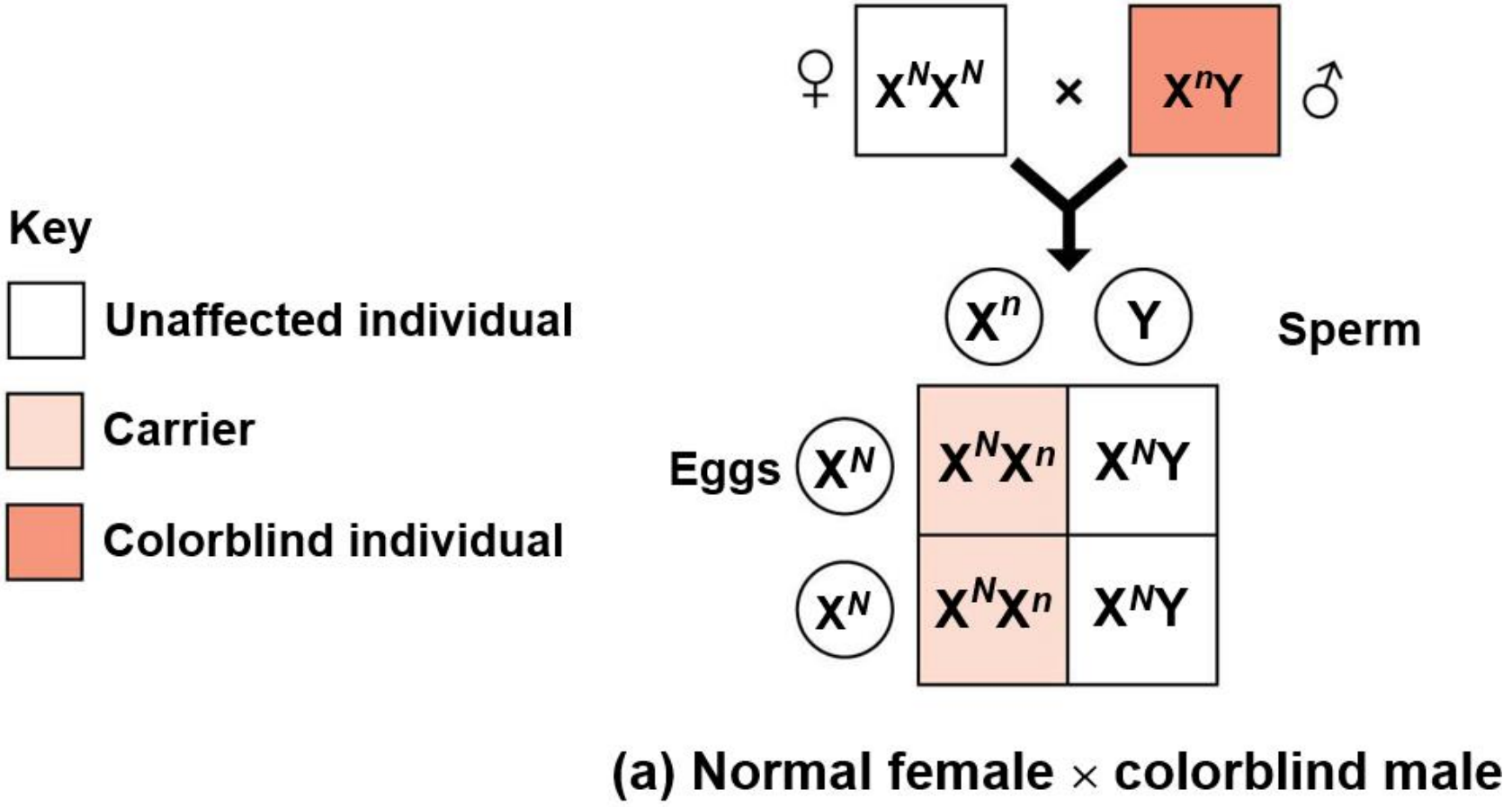
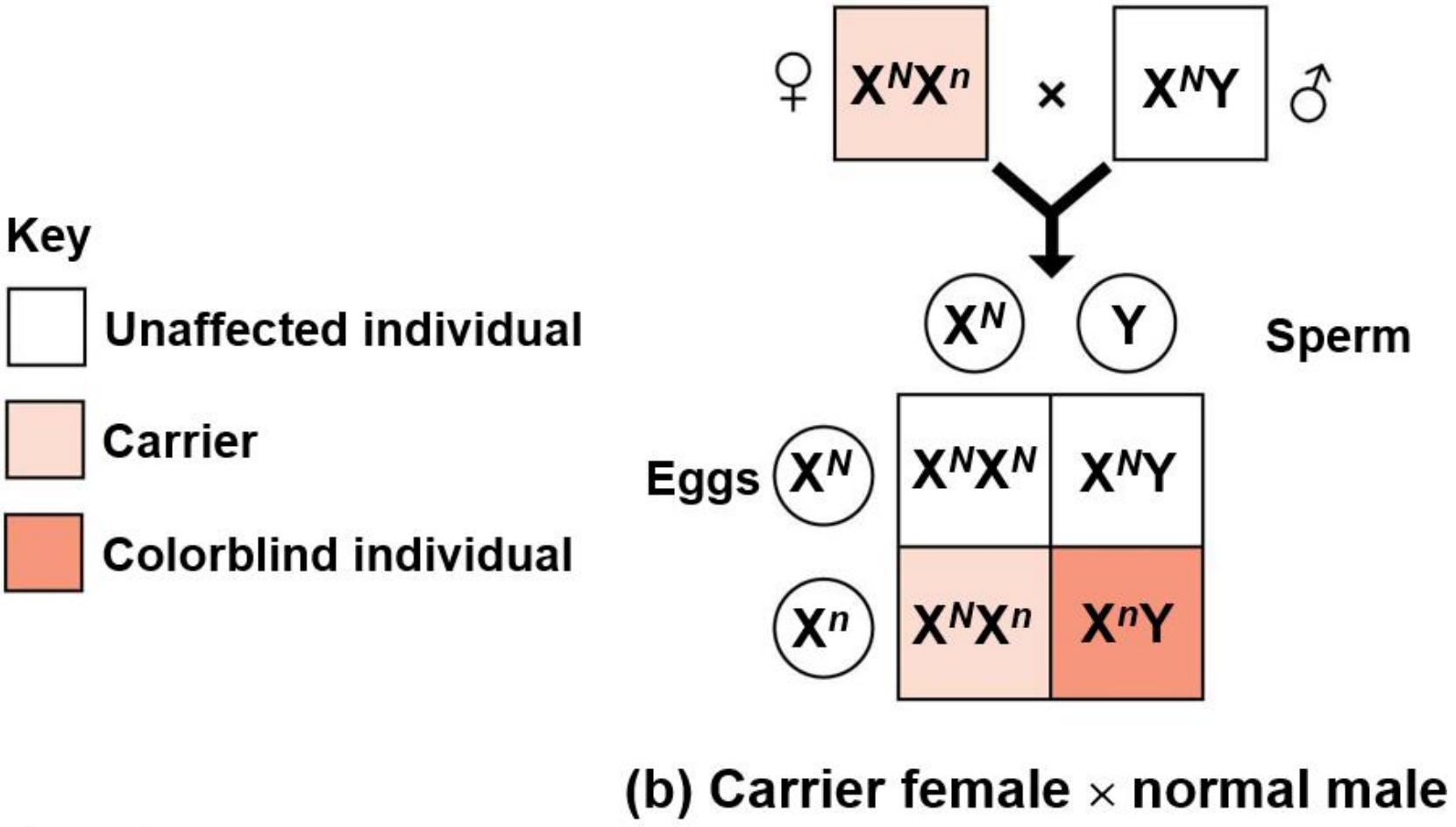
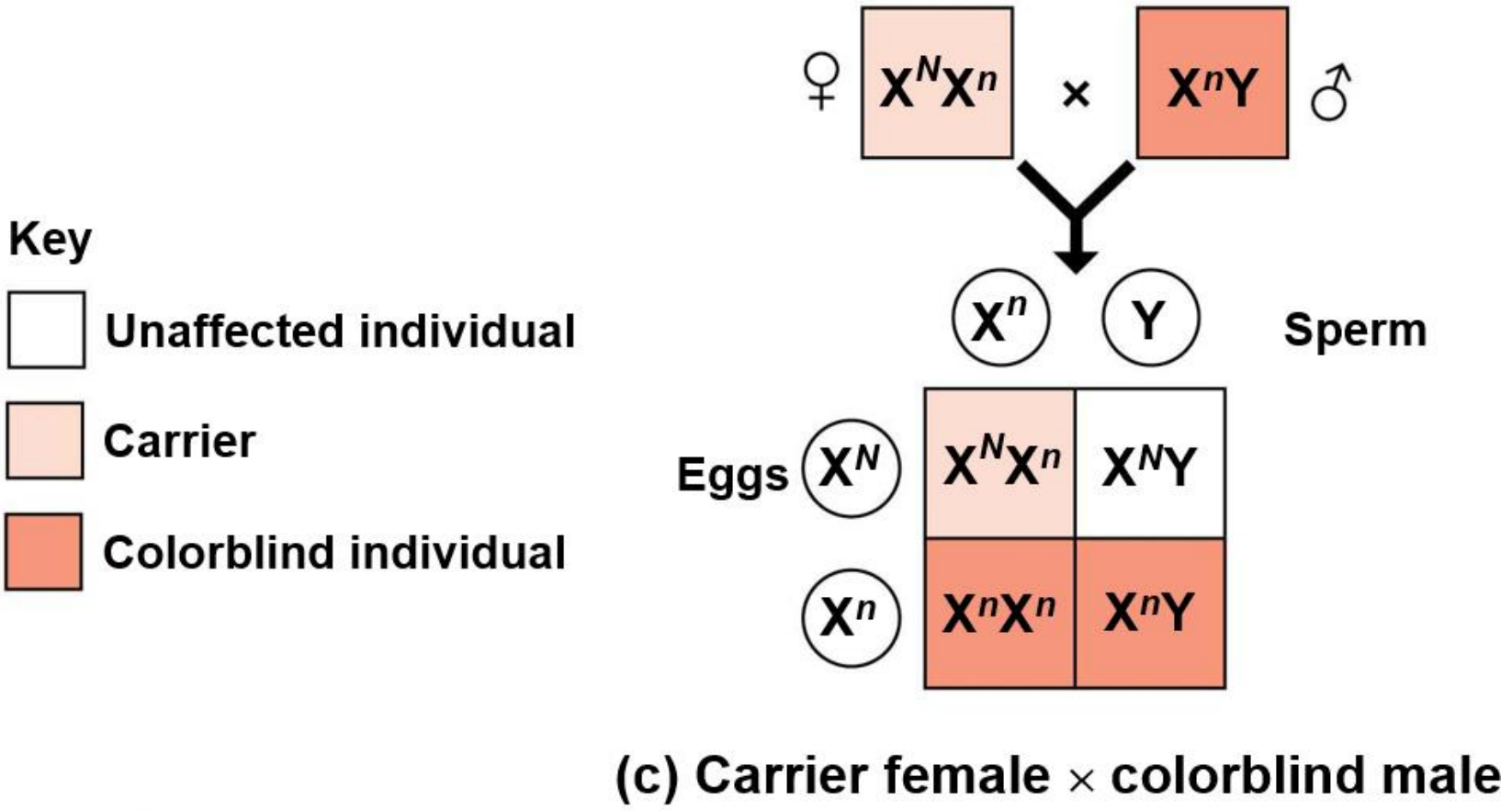


Figure 9.27 Inheritance of Colorblindness, a Sex-Linked Recessive Trait (3 of 4)



(b) Carrier female × normal male

Figure 9.27 Inheritance of Colorblindness, a Sex-Linked Recessive Trait (4 of 4)



(c) Carrier female × colorblind male

Sex-Linked Genes (5 of 5)

- Hemophilia is a sex-linked recessive trait with a long, well-documented history.
  - Hemophiliacs bleed excessively when injured because they have inherited an abnormal allele for a factor involved in blood clotting.
  - The most seriously affected individuals may bleed to death after relatively minor bruises or cuts.
  - The age-old practice of strengthening international alliances by marriage effectively spread hemophilia through the royal families of several nations.

Figure 9.28 Hemophilia in the Royal Family of Russia (1 of 3)

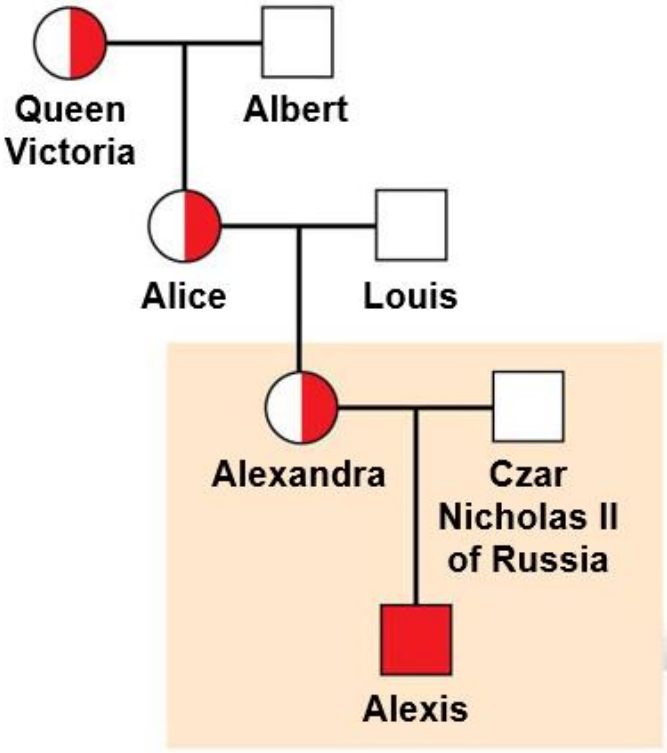


Figure 9.28 Hemophilia in the Royal Family of Russia (2 of 3)

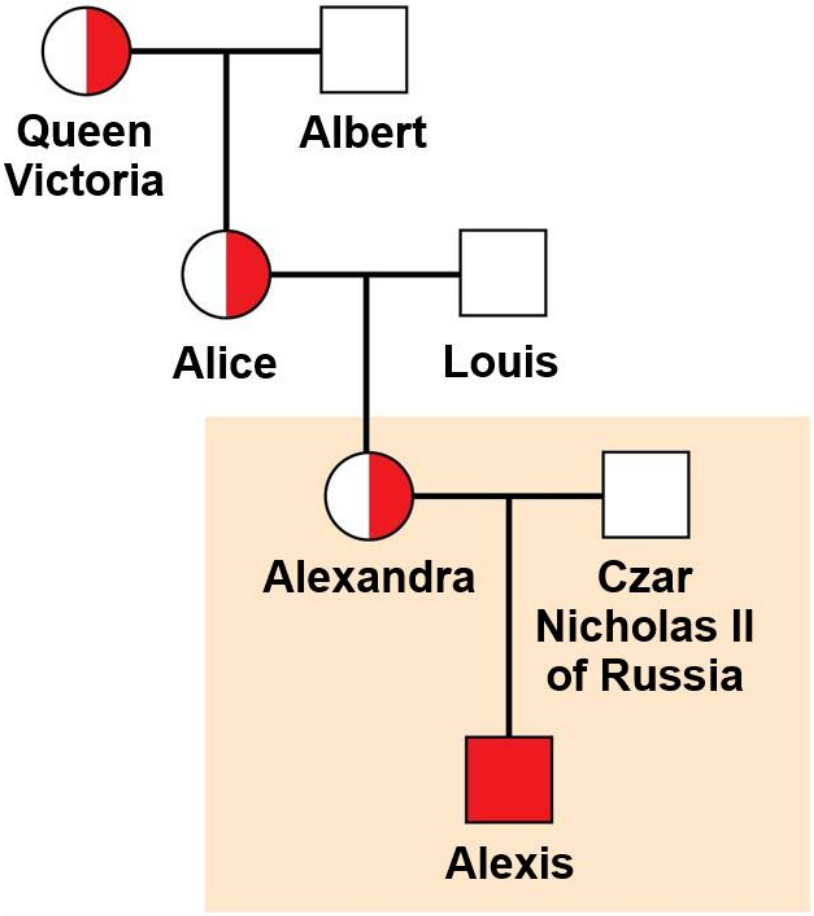


Figure 9.28 Hemophilia in the Royal Family of Russia (3 of 3)

Evolution Connection: Barking Up the Evolutionary Tree (1 of 3)

- About 15,000 years ago, in East Asia, people began to cohabit with ancestral canines that were predecessors of both modern wolves and dogs.
- As people moved into permanent, geographically isolated settlements, populations of canines were separated from one another and eventually became inbred.

Evolution Connection: Barking Up the Evolutionary Tree (2 of 3)

- A 2010 study indicated that small dogs were first bred within early agricultural settlements of the Middle East around 12,000 years ago.
- Continued over millennia, such genetic tinkering has resulted in a diverse array of dog body types and behaviors.

Evolution Connection: Barking Up the Evolutionary Tree (3 of 3)

- Our understanding of canine evolution took a big leap forward when researchers sequenced the complete genome of a dog.
- Using the genome sequence and a wealth of other data, canine geneticists produced an evolutionary tree based on a genetic analysis of 85 breeds.
- The formulation of an evolutionary tree for the domestic dog shows that new technologies can provide important insights into genetic and evolutionary questions about life on Earth.

Figure 9.29 An Evolutionary Tree of Dog Breeds (1 of 2)

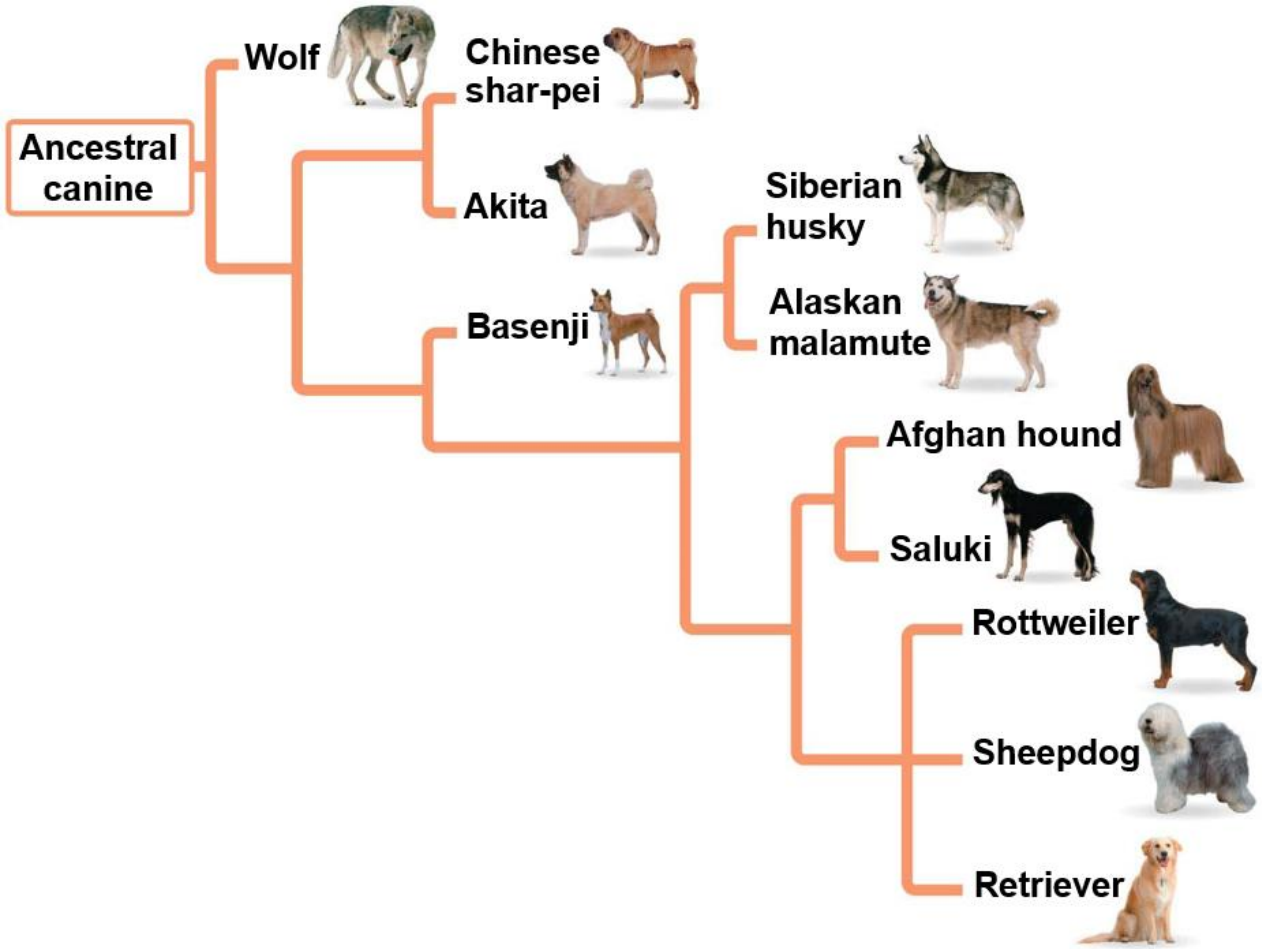


Figure 9.29 An Evolutionary Tree of Dog Breeds (2 of 2)

Figure 9.UN01

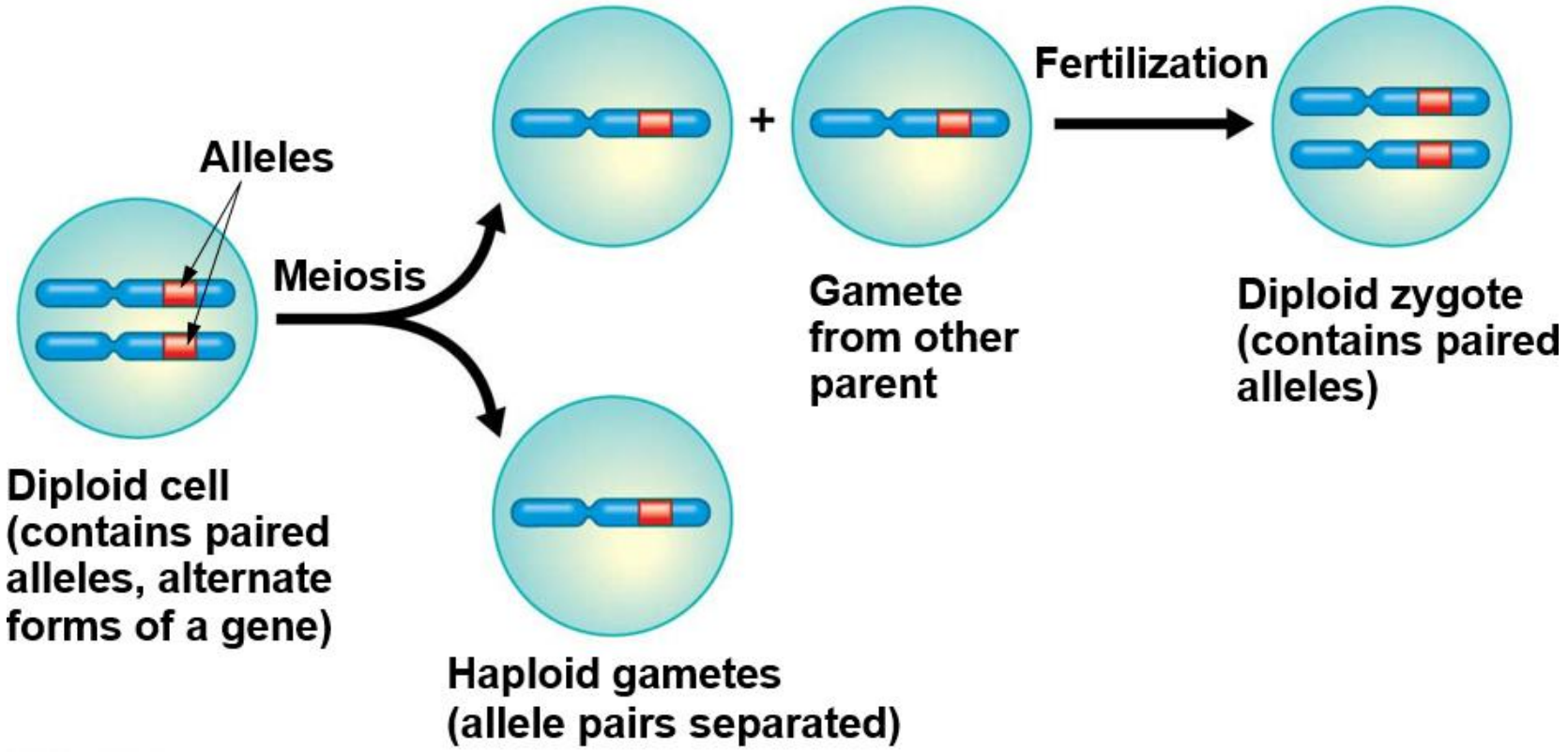


Figure 9.UN02

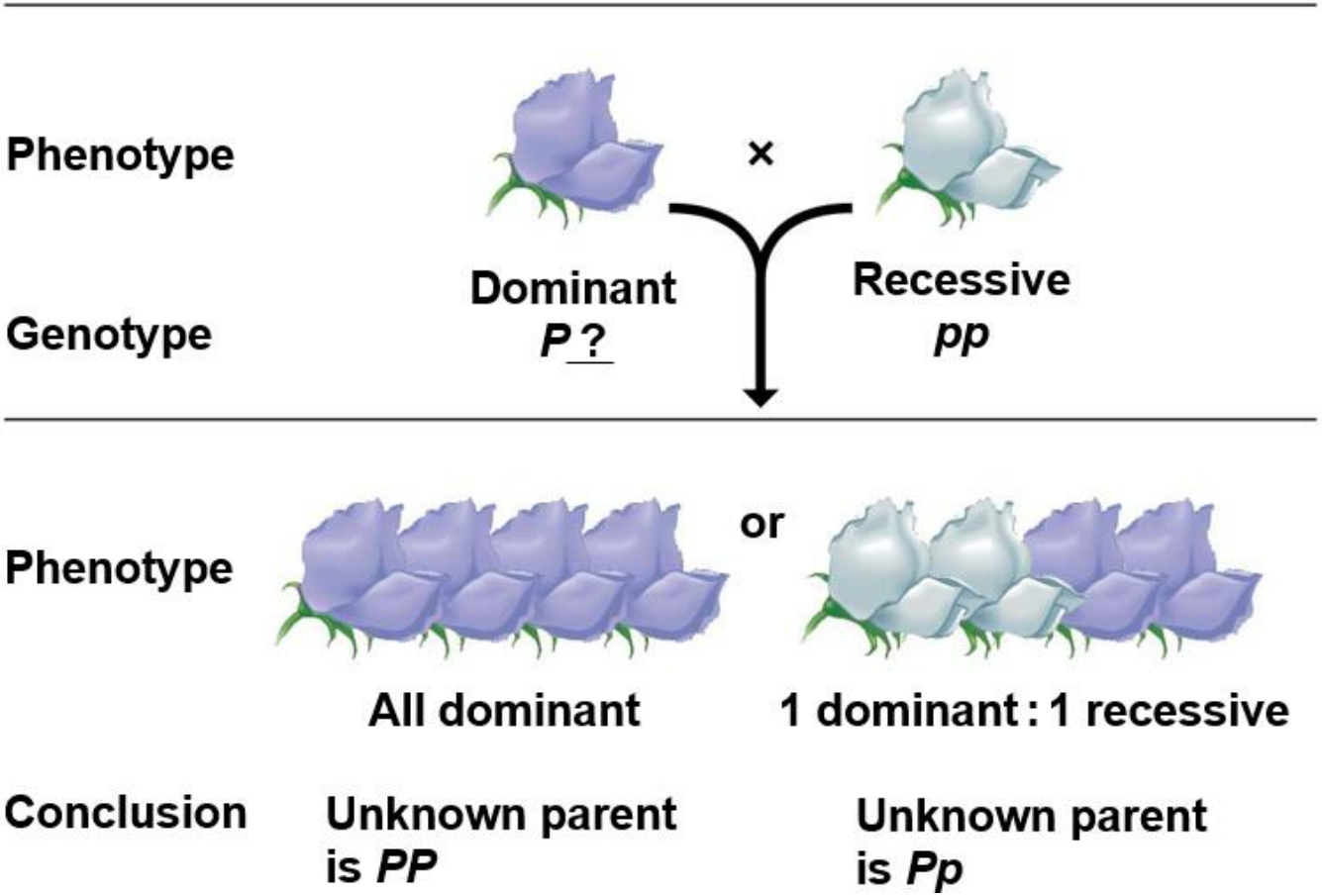


Figure 9.UN03

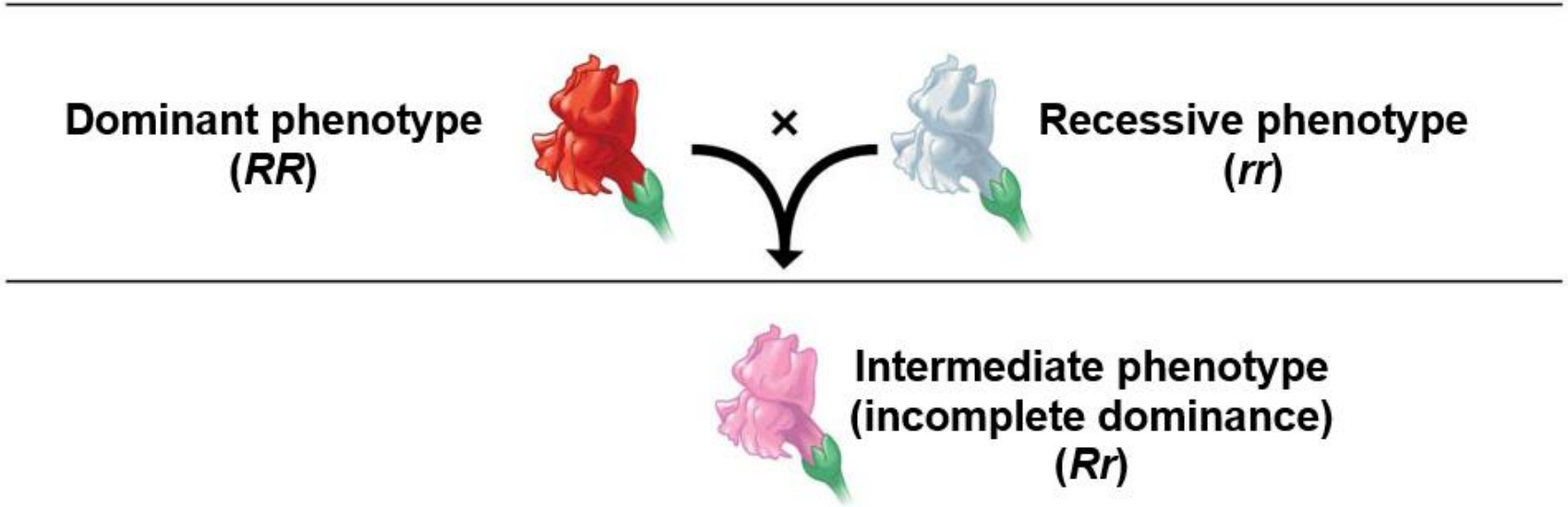


Figure 9.UN04

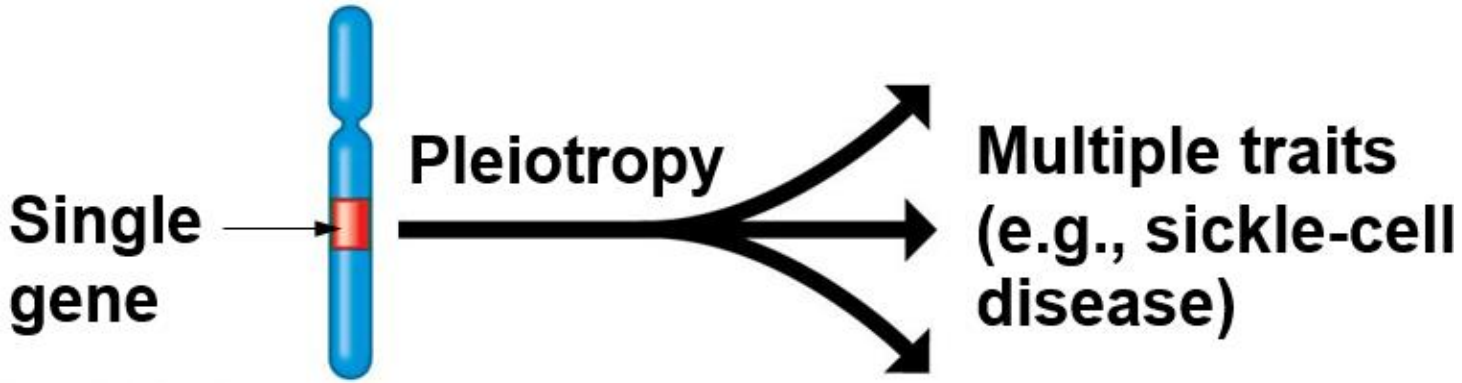


Figure 9.UN05

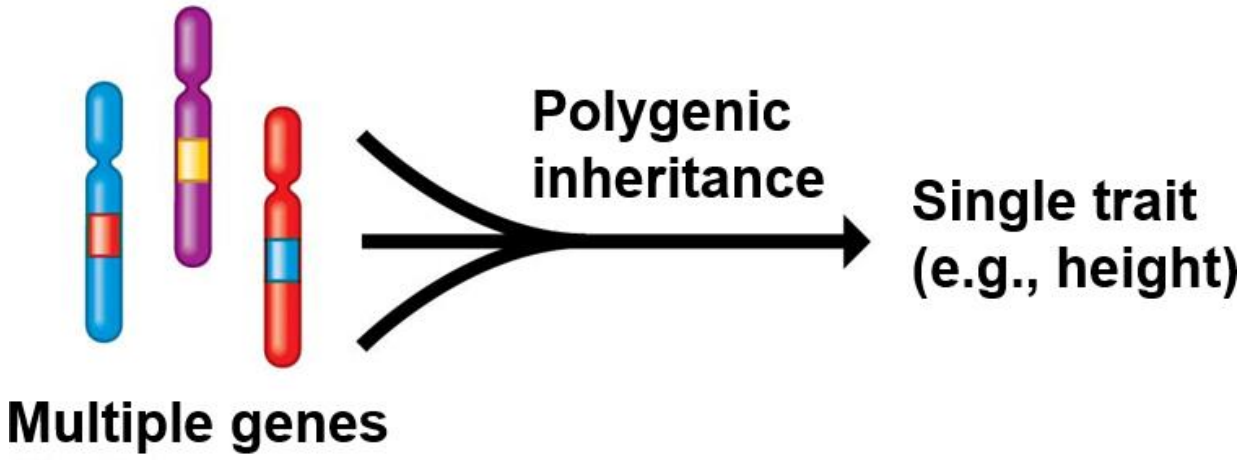


Figure 9.UN06

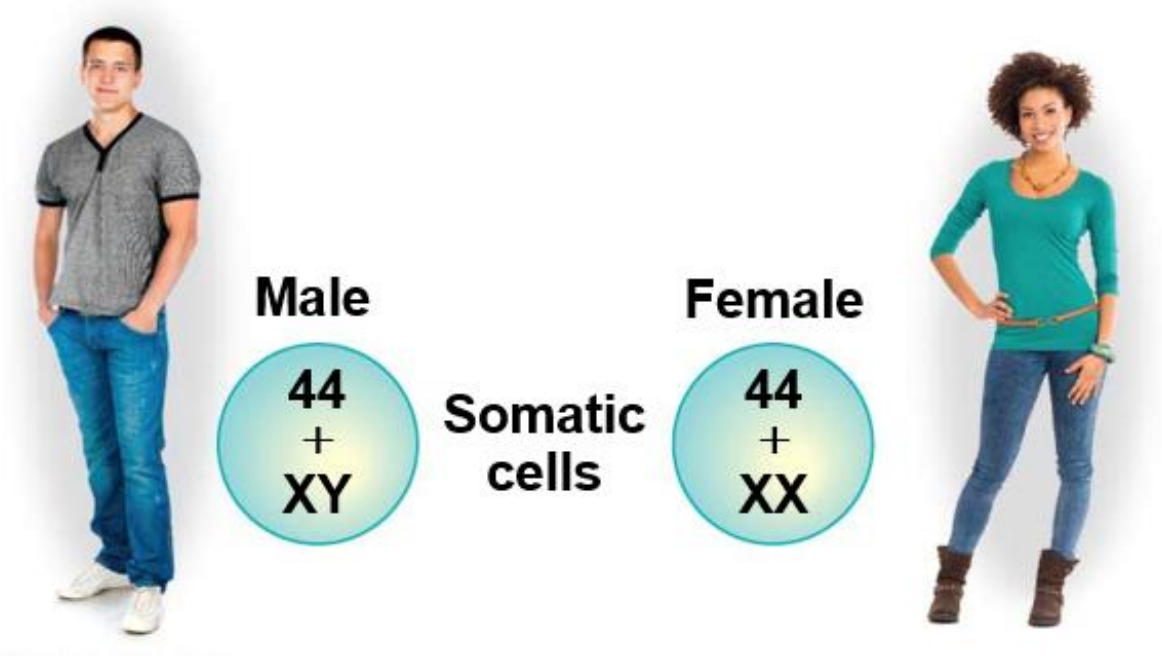


Figure 9.UN07

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.UN08

Figure 9.UN09

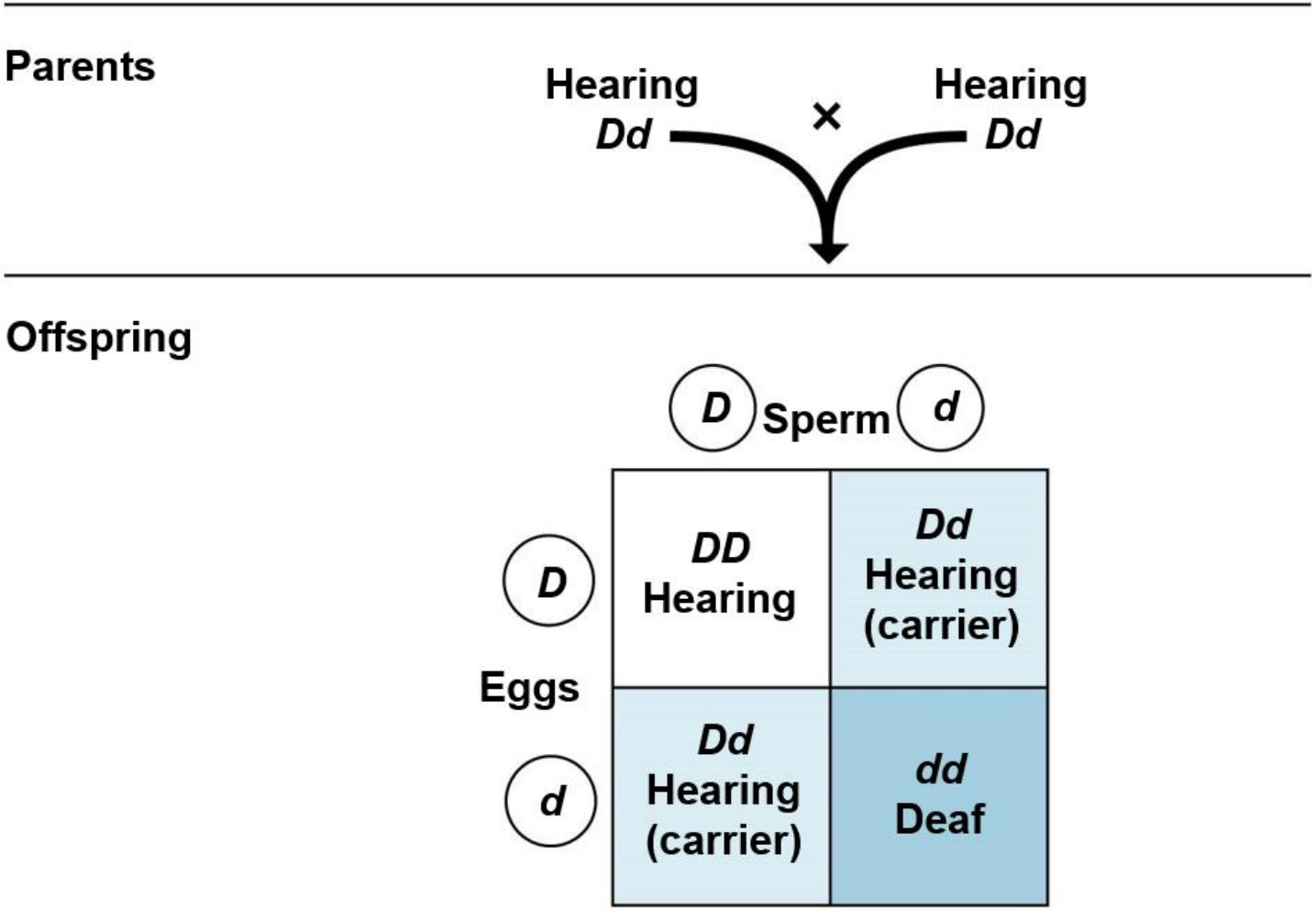


Figure 9.UN10