Mendel's Laws

9.1 The Study of Genetics Has Ancient Roots (1 of 2)

- Hippocrates's idea that "pangenes" travel from each part of an organism's body to the eggs or sperm is incorrect because
  - the reproductive cells are not composed of particles from somatic (body) cells and
  - changes in somatic cells do not influence gametes.
- The blending hypothesis was rejected because it did not explain how traits that disappear in one generation can reappear in later generations.

9.1 The Study of Genetics Has Ancient Roots (2 of 2)

Checkpoint question Imagine you have two different birds of the same species, a female with a yellow beak and a male with a blue beak. Design a simple experiment to test the blending hypothesis.

Cross the two birds and observe the resulting beak color in the offspring. The blending hypothesis predicts the appearance of green beaks.

9.2 The Science of Genetics Began in an Abbey Garden (1 of 2)

- **Heredity** is the transmission of traits from one generation to the next.
- **Genetics (**the scientific study of heredity) began with Gregor Mendel's experiments.
  - A heritable feature that varies among individuals is called a **character**.
  - Mendel crossed pea plants and traced **traits** (variants of a character) from generation to generation.
  - He hypothesized that there are alternative versions of genes (alleles), the units that determine heritable traits.

9.2 The Science of Genetics Began in an Abbey Garden (2 of 2)

Checkpoint question Describe the three generations of your own family using the terminology of a genetic cross (P, F_1, F_2).

The P generation is your grandparents, the F_1 your parents, and the F_2 is you (and any siblings).

Figure 9.2c_3

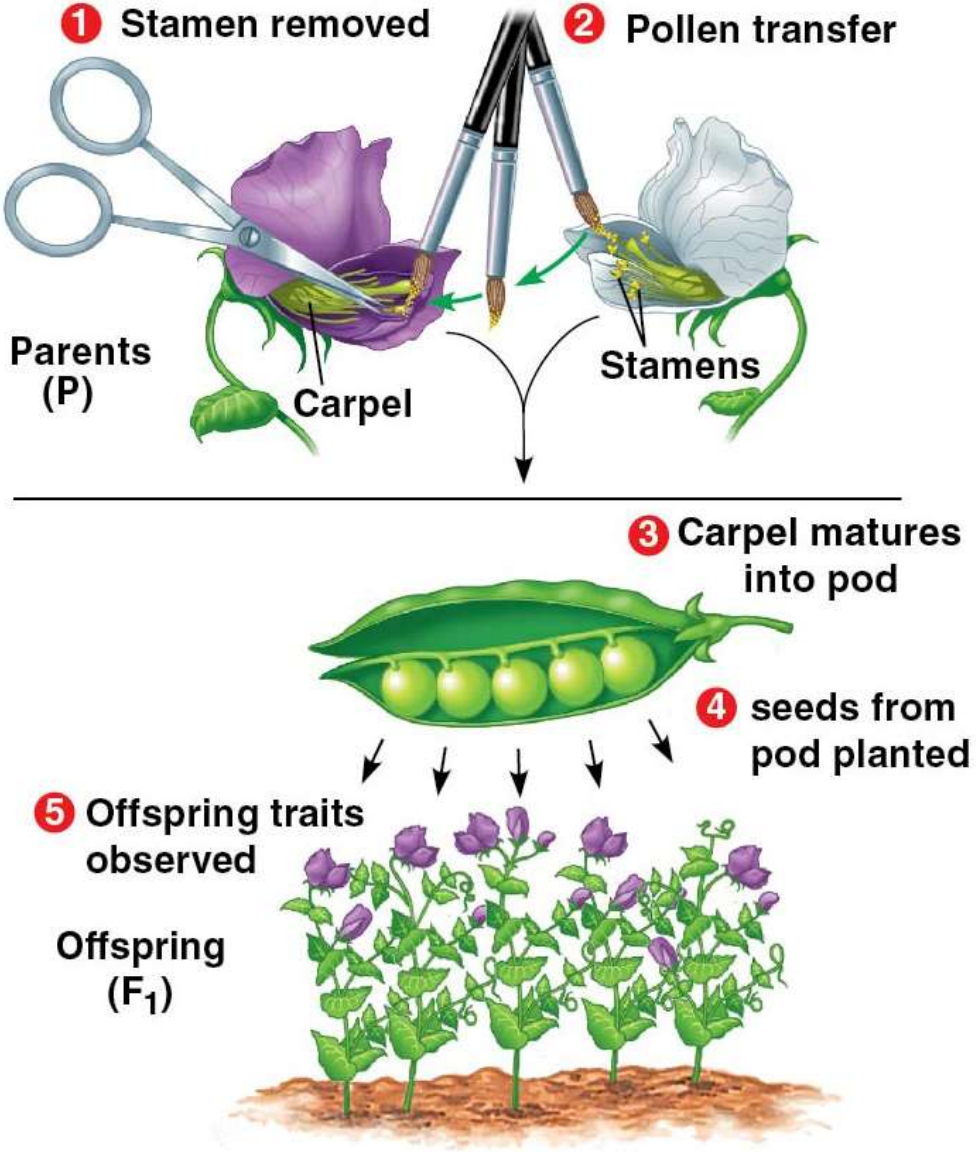


Figure 9.2d

Character	Traits	
	Dominant	Recessive
Flower color	 Purple	 White
Flower position	 Axial	 Terminal
Seed color	 Yellow	 Green
Seed shape	 Round	 Wrinkled
Pod shape	 Inflated	 Constricted
Pod color	 Green	 Yellow
Stem length	 Tall	 Dwarf

9.3 Mendel's Law of Segregation Describes the Inheritance of a Single Character (1 of 3)

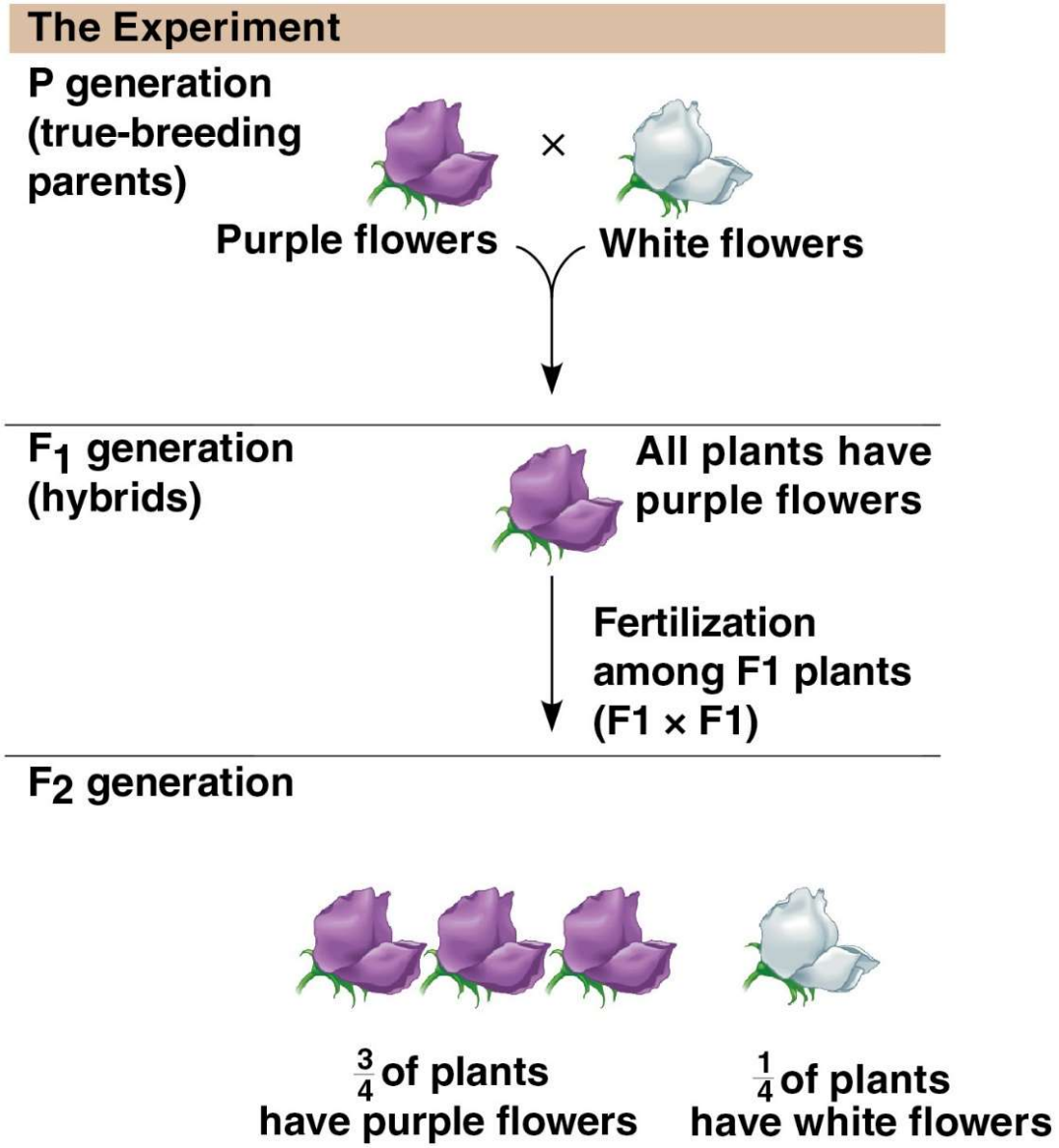
- Mendel developed four hypotheses, described below using modern terminology.

1. *There are alternative versions of genes* (called **alleles**) *that account for variations in inherited characters.*
2. *For each character, an organism inherits two alleles of a gene, 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.

9.3 Mendel's Law of Segregation Describes the Inheritance of a Single Character (2 of 3)

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**.
4. *A sperm or egg carries only one allele for each inherited character because allele pairs separate (segregate) from each other during the production of gametes.* This statement is called the **law of segregation**.

Figure 9.3a_3



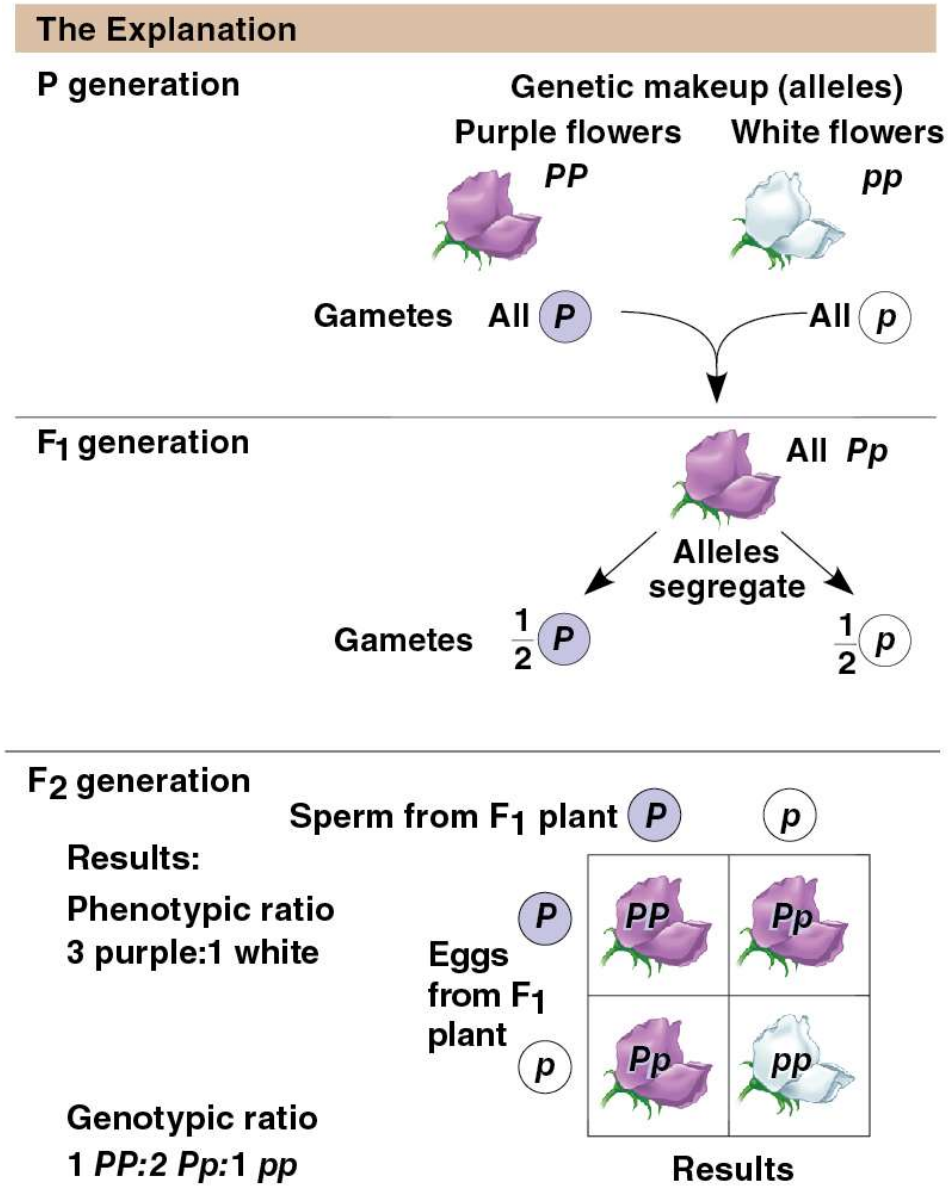
9.3 Mendel's Law of Segregation Describes the Inheritance of a Single Character (3 of 3)

- Mendel's hypotheses also explain the 3:1 ratio observed in the F_2 generation.
  - The F_1 hybrids all have a *Pp* **genotype**.
  - A **Punnett square** shows the four possible combinations of alleles that could occur when these gametes combine.

Checkpoint question How can two plants with different genotypes for a particular inherited character be identical in phenotype?

One could be homozygous for the dominant allele and the other heterozygous.

Figure 9.3b_3



9.4 Homologous Chromosomes Bear the Alleles for Each Character

- Every diploid cell has pairs of homologous chromosomes.
- The chromosomes in a homologous pair carry alleles of the same genes at the same locations.

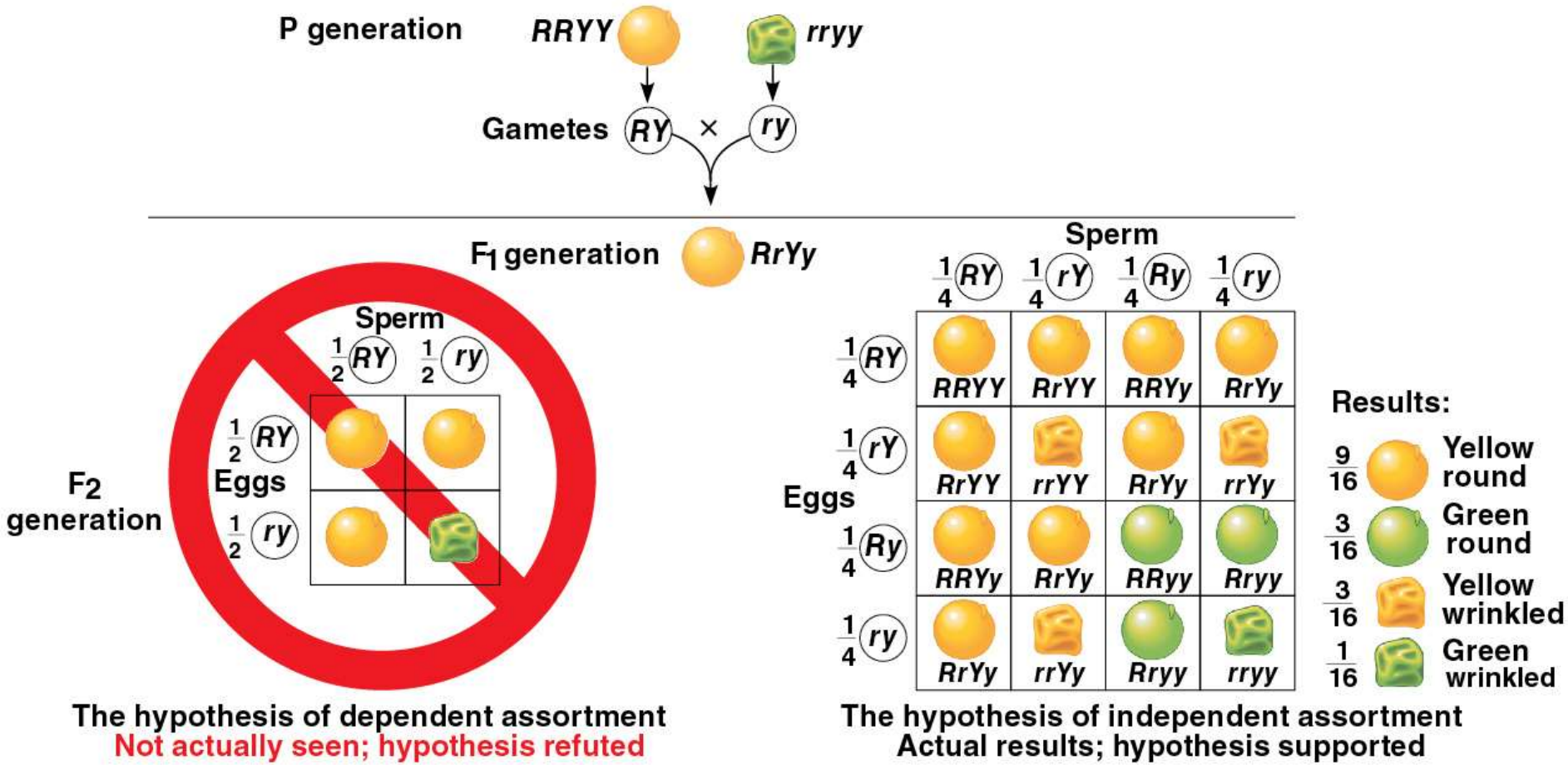
Checkpoint question An individual is heterozygous, *Bb*, for a gene. According to the law of segregation, each gamete formed by this individual will have either the *B* allele or the *b* allele. Which step in the process of meiosis is the physical basis for this segregation of alleles?

The *B* and *b* alleles are located at the same gene locus on homologous chromosomes, which separate during meiosis I and are packaged in separate gametes during meiosis II.

9.5 The Law of Independent Assortment Is Revealed by Tracking Two Characters at Once (1 of 2)

- A cross between two individuals that are heterozygous for one character is called a **monohybrid cross**.
- A **dihybrid cross** is a cross between two organisms that are each heterozygous for two characters being followed
- Mendel's **law of independent assortment** states that the alleles of a pair segregate independently of other allele pairs during gamete formation.

Figure 9.5a



9.5 The Law of Independent Assortment Is Revealed by Tracking Two Characters at Once (2 of 2)

Checkpoint question Predict the phenotypes of offspring obtained by mating a black Lab homozygous for both coat color and normal eyes with a chocolate Lab that is blind from PRA.

All offspring would be black with normal eyes ($BBNN \times bbnn \rightarrow BbNn$).

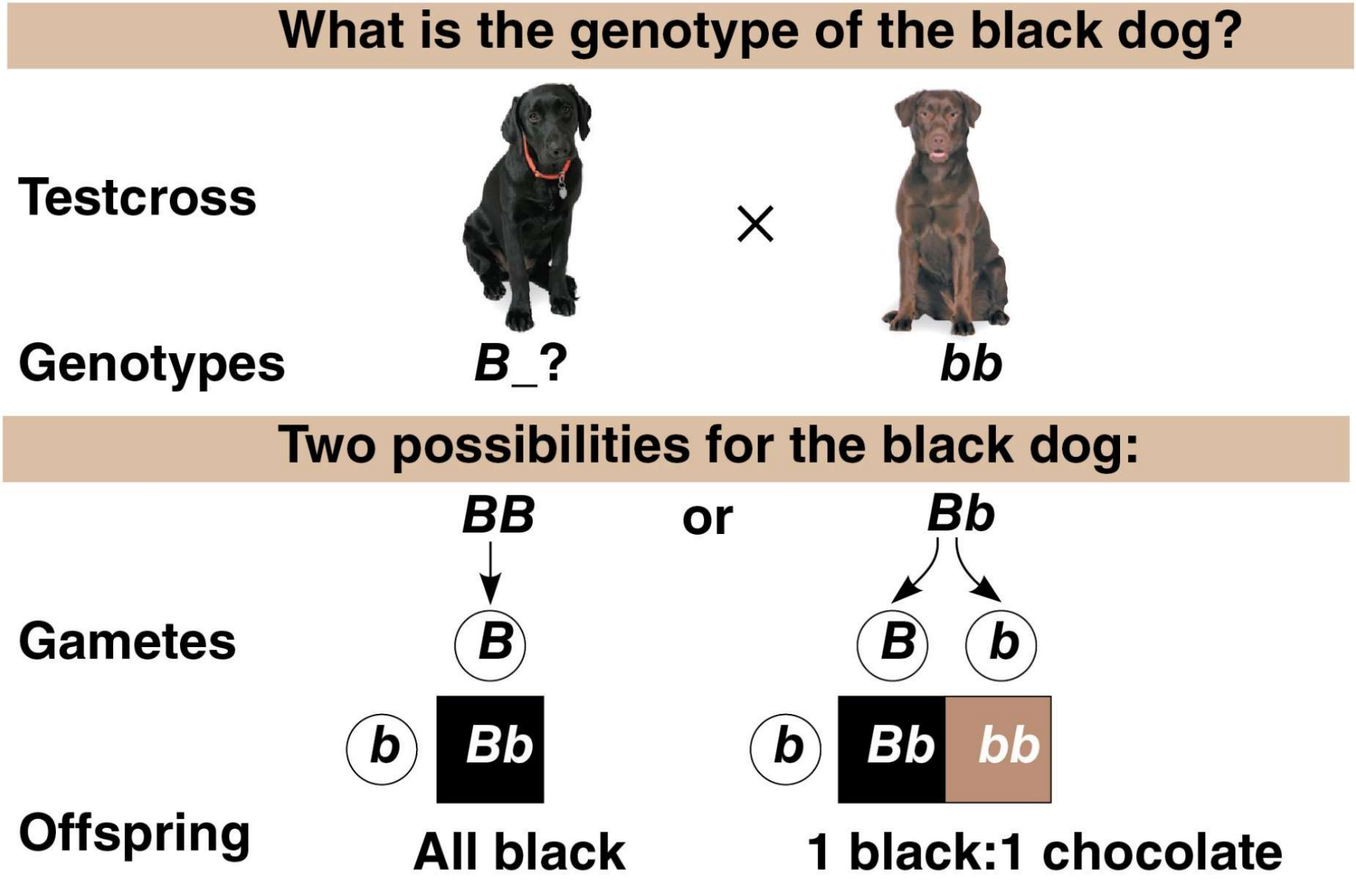
9.6 Geneticists Can Use a Testcross to Determine Unknown Genotypes

- The offspring of a **testcross**, a mating between an individual of unknown genotype and a homozygous recessive individual, can reveal the unknown genotype.

Checkpoint question You use a testcross to determine the genotype of a Lab with normal eyes. Half of the offspring are normal and half develop PRA. What is the genotype of the normal parent?

Heterozygous (*Nn*)

Figure 9.6



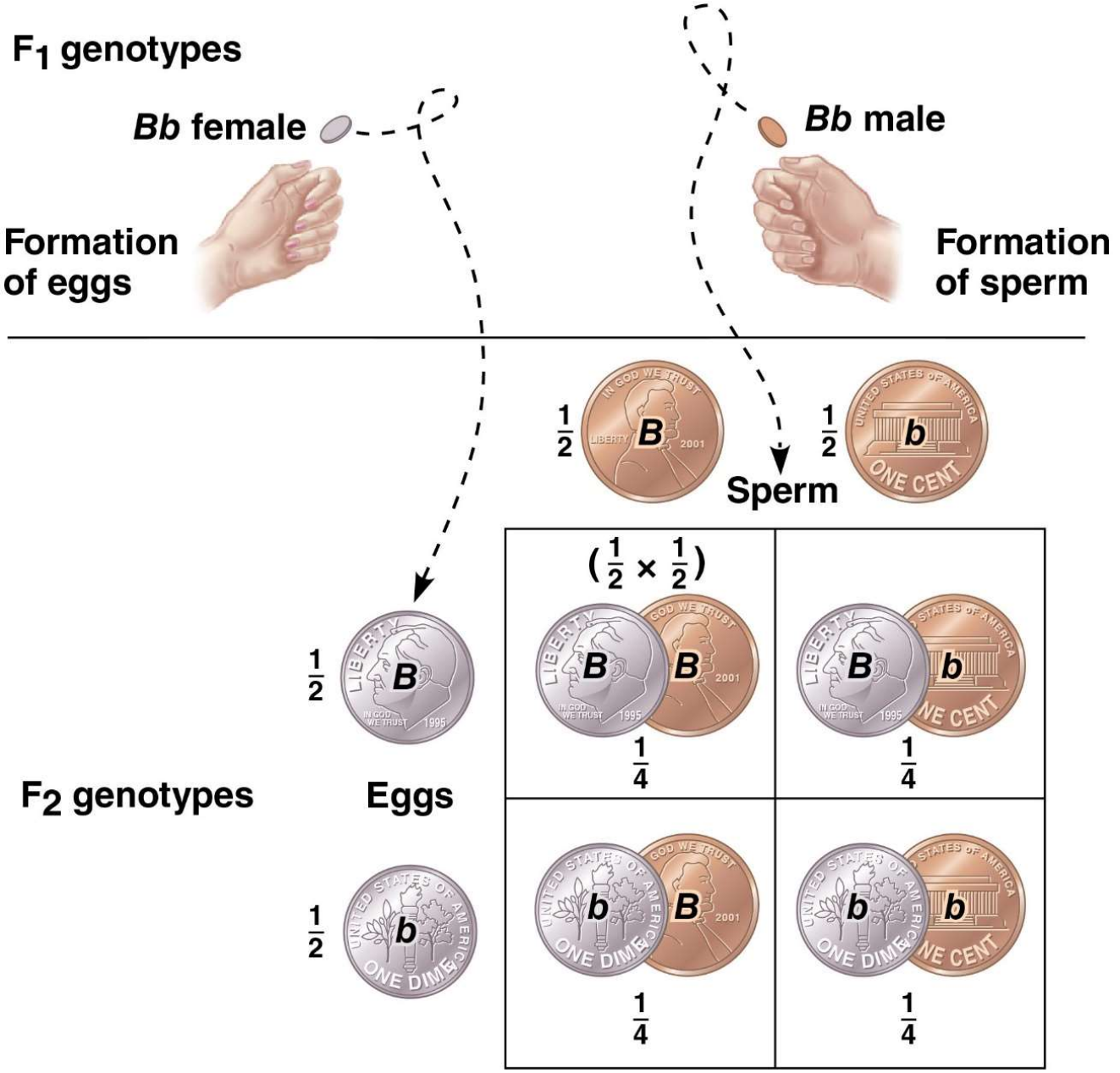
9.7 Mendel's Laws Reflect the Rules of Probability

- The **rule of multiplication** calculates the probability of two independent events both occurring.
- The **rule of addition** calculates the probability of an event that can occur in alternative ways.

Checkpoint question A plant of genotype *AABbCC* is crossed with an *AaBbCc* plant. What is the probability of an offspring having the genotype *AABBCC*? (Hint: Treat this as 3 separate monohybrid crosses.)

Heterozygous (*Nn*)

Figure 9.7



9.8 Visualizing the Concept: Genetic Traits in Humans Can Be Tracked Through Family Pedigrees

- The inheritance of many human traits follows Mendel's laws.
- Family **pedigrees (a family tree)** can help determine individual genotypes.

Checkpoint question If Libby had a child, which phenotype would allow her to deduce her own genotype for certain?

If her child had a straight hairline (*hh*), then
Libby would know that she herself must be *Hh*.

9.9 Connection: Many Inherited Traits in Humans Are Controlled by a Single Gene (1 of 2)

- The genetic disorders listed in Table 9.9 are known to be inherited as dominant or recessive traits controlled by a single gene.
- Most people who have recessive disorders are born to normal parents who are both heterozygotes—that is, parents who are **carriers** of the recessive allele for the disorder but are phenotypically normal.

Table 9.9 Some Autosomal Disorders in Humans

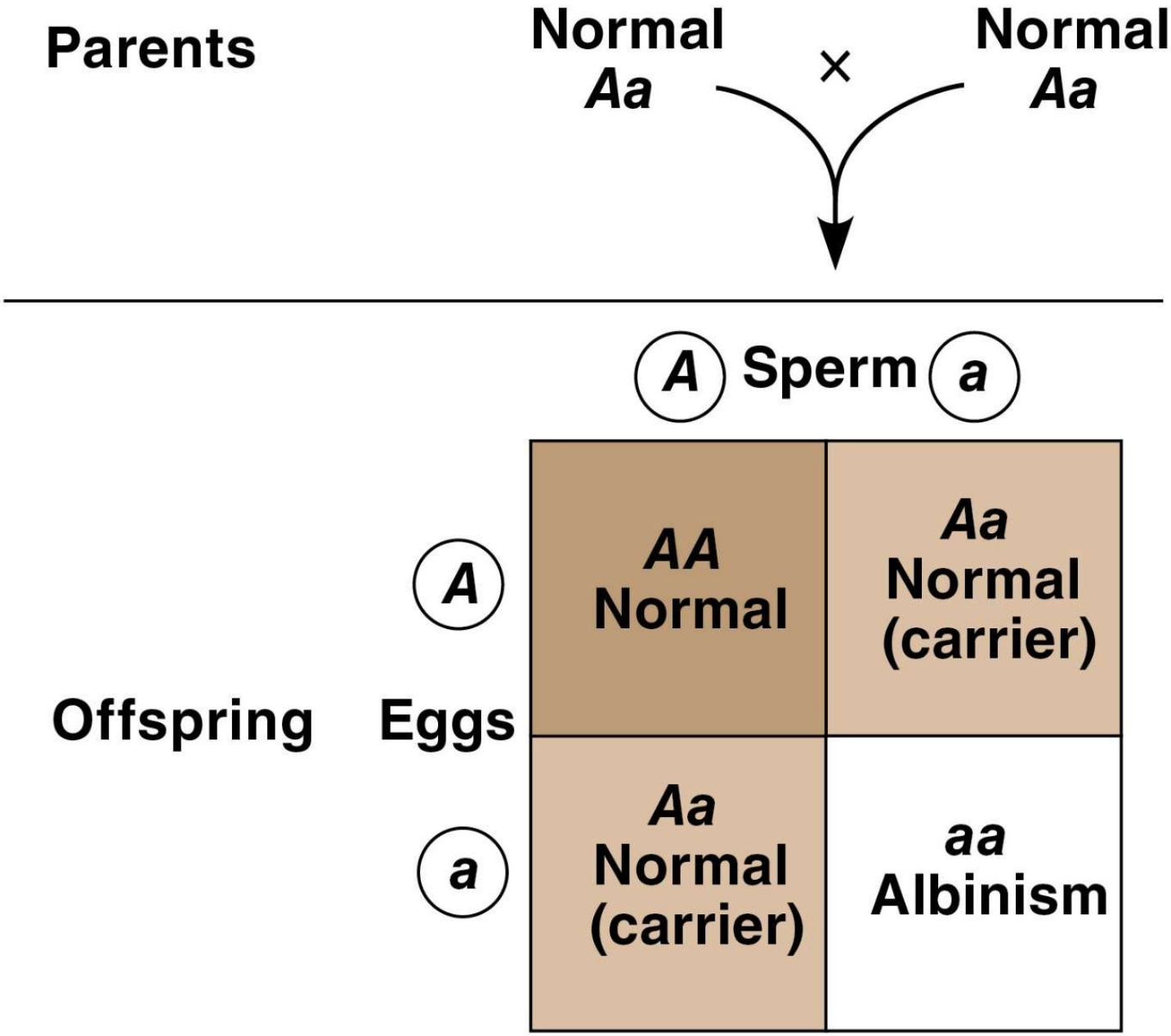
Disorder	Major Symptoms
Recessive Disorders	
Albinism	Lack of pigment in the skin, hair, and eyes
Cystic fibrosis	Excess mucus in the lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; developmental disabilities
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
Huntington's disease	Uncontrollable movements; cognitive impairments; strikes in middle age
Hypercholesterolemia	Excess cholesterol in the blood; heart disease

9.9 Connection: Many Inherited Traits in Humans Are Controlled by a Single Gene (2 of 2)

Checkpoint question Peter is a 30-year-old man whose father died of Huntington's disease. Neither Peter's mother nor a much older sister shows any signs of Huntington's. What is the probability that Peter has inherited Huntington's disease?

Since his father had the disease, there is a 1/2 chance that Peter received the gene. (The genotype of his sister is irrelevant.)

Figure 9.9b



9.10 Connection: New Technologies Can Provide Insight into One's Genetic Legacy

- Carrier screening, fetal testing, fetal imaging, and newborn screening can provide information for reproductive decisions but may create ethical dilemmas.

Checkpoint question What is the primary benefit of genetic screening by CVS? What is the primary risk?

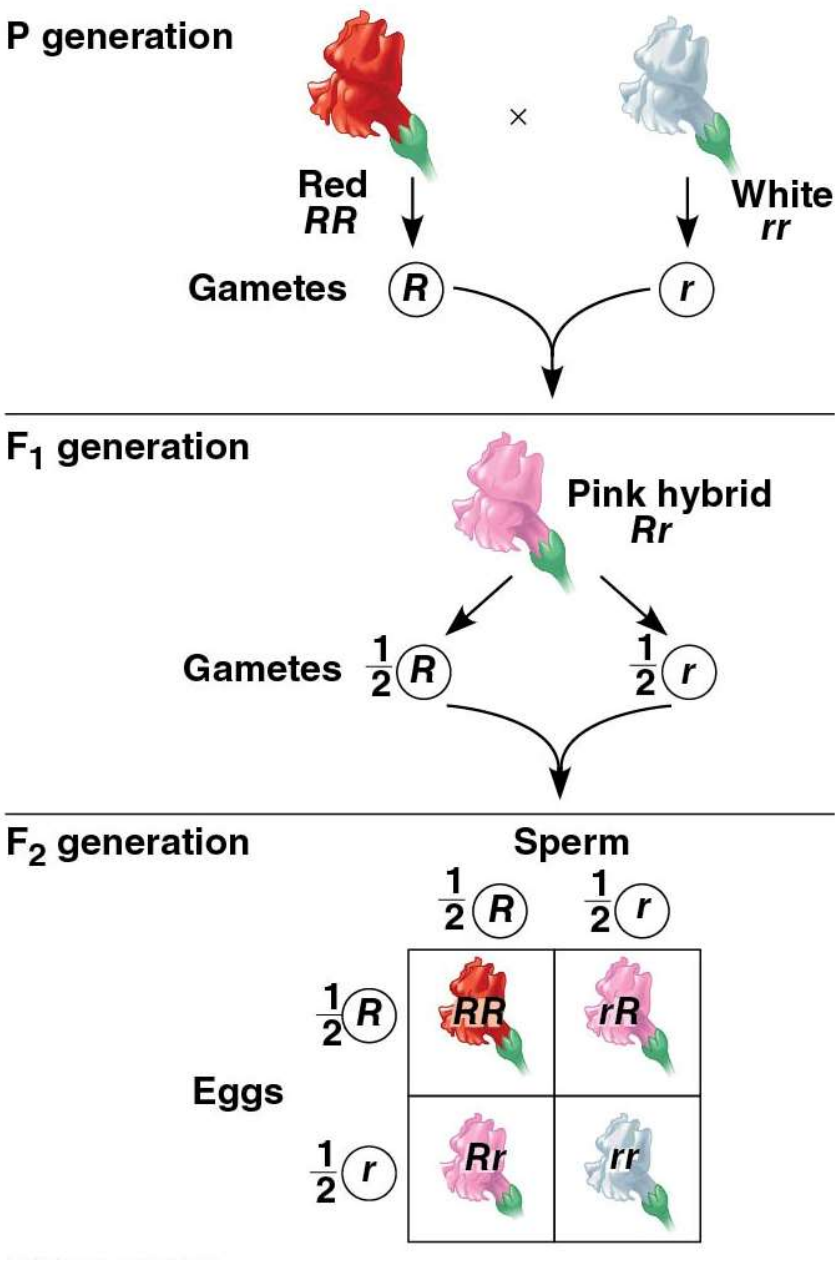
CVS allows genetic screening to be performed very early in pregnancy and provides quick results, but it carries a risk of miscarriage.

Variations on Mendel's Laws

9.11 Incomplete Dominance Results in Intermediate Phenotypes (1 of 2)

- Mendel's laws are valid for all sexually reproducing species, but genotype often does not dictate phenotype in the simple way Mendel's laws describe.
  - Mendel's pea crosses always looked like one of the two parental varieties, a situation called **complete dominance**.
  - For some characters, the appearance of F_1 hybrids falls between the phenotypes of the two parental varieties. This is called **incomplete dominance**.

Figure 9.11a



9.11 Incomplete Dominance Results in Intermediate Phenotypes (2 of 2)

Checkpoint question Why doesn't the cross shown in Figure 9.11A support the blending hypothesis?

Although two of the F_2 offspring show a "blended" phenotype (pink flowers), the other two do not, and the white and red alleles are not lost to future generations.

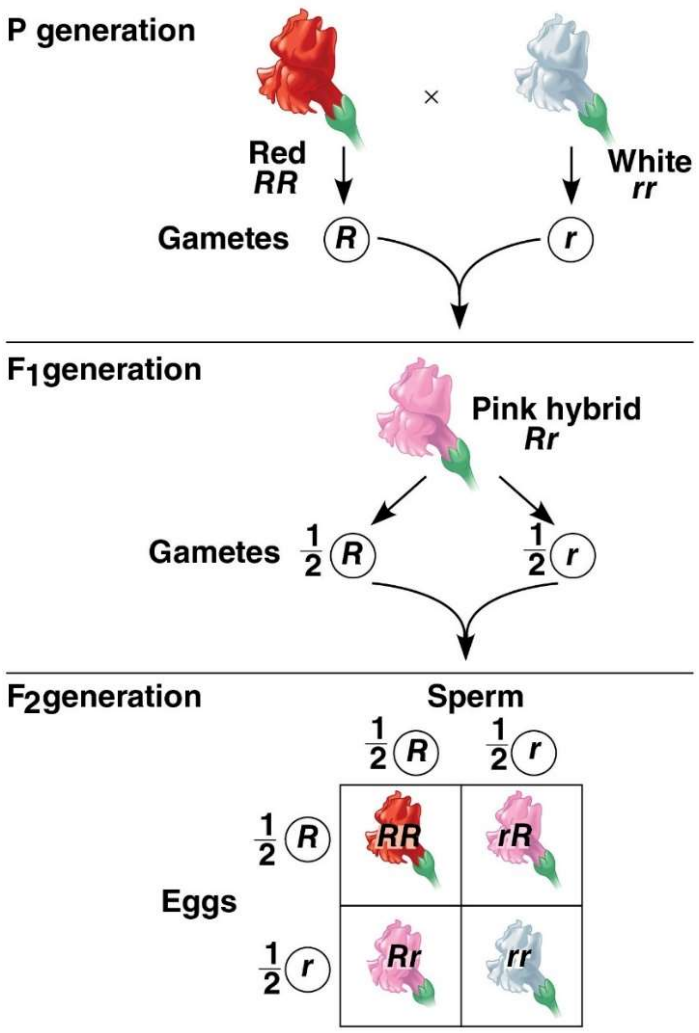
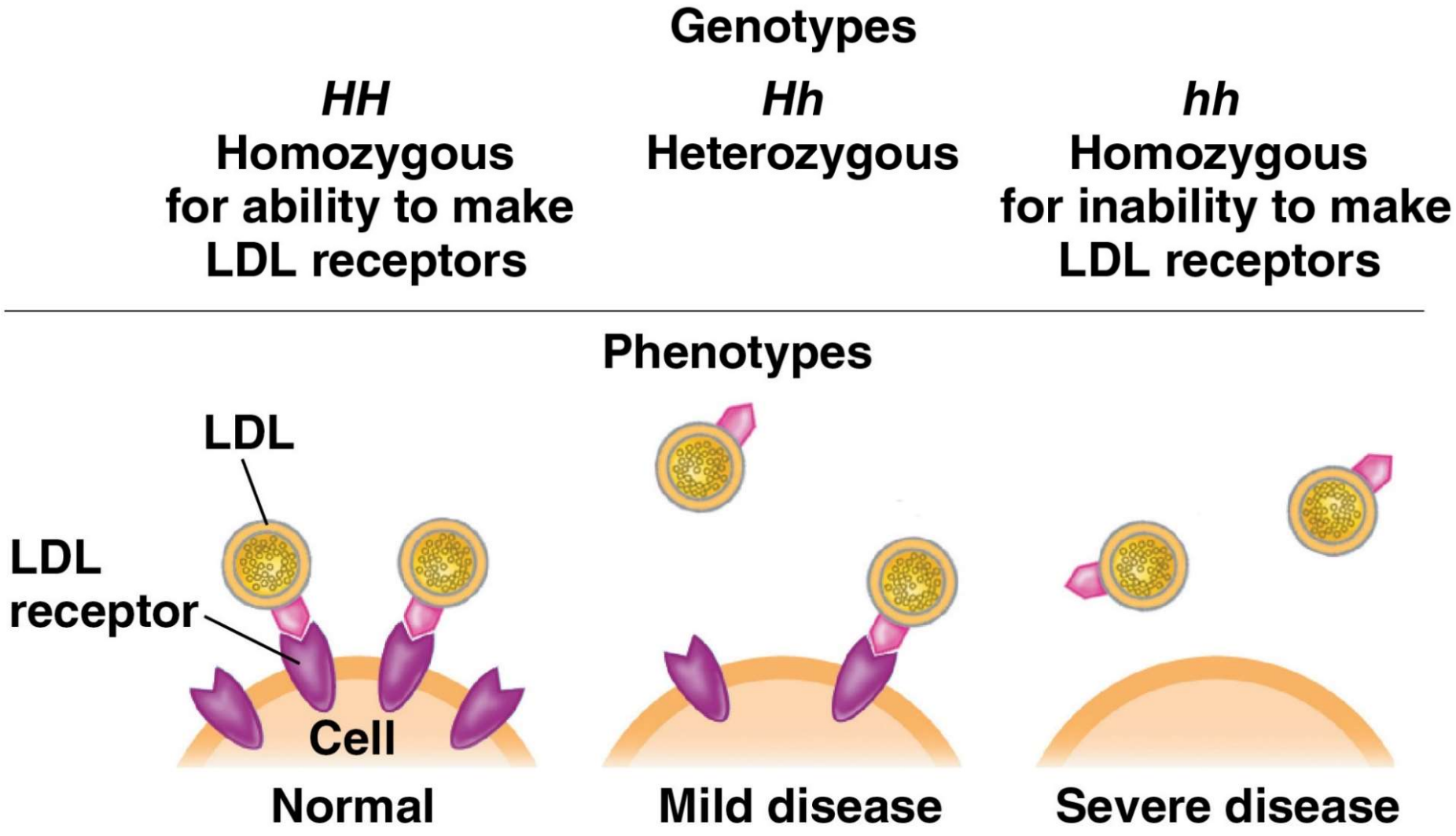


Figure 9.11b



9.12 Many Genes Have More Than Two Alleles That May Be Codominant (1 of 2)

- The **ABO blood group** phenotype in humans is controlled by three alleles that produce a total of four phenotypes.
- The I^A and I^B alleles are **codominant**: Both alleles are expressed in heterozygous individuals ($I^A I^B$), who have type AB blood.

9.12 Many Genes Have More Than Two Alleles That May Be Codominant (2 of 2)

Checkpoint question Maria has type O blood, and her sister has type AB blood. The girls know that both of their maternal grandparents are type A. What are the genotypes of the girls' parents?

Their mother is $I^A i$ and their father is $I^B i$.

Figure 9.12

Blood Group (Phenotype)	Genotypes	Carbohydrates Present on Red Blood Cells	Antibodies Present in Blood	Reaction 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	$I^A I^B$	Carbohydrate A and Carbohydrate B	None				
O	ii	Neither	Anti-A Anti-B				

9.13 A Single Gene May Affect Many Phenotypic Characters (1 of 2)

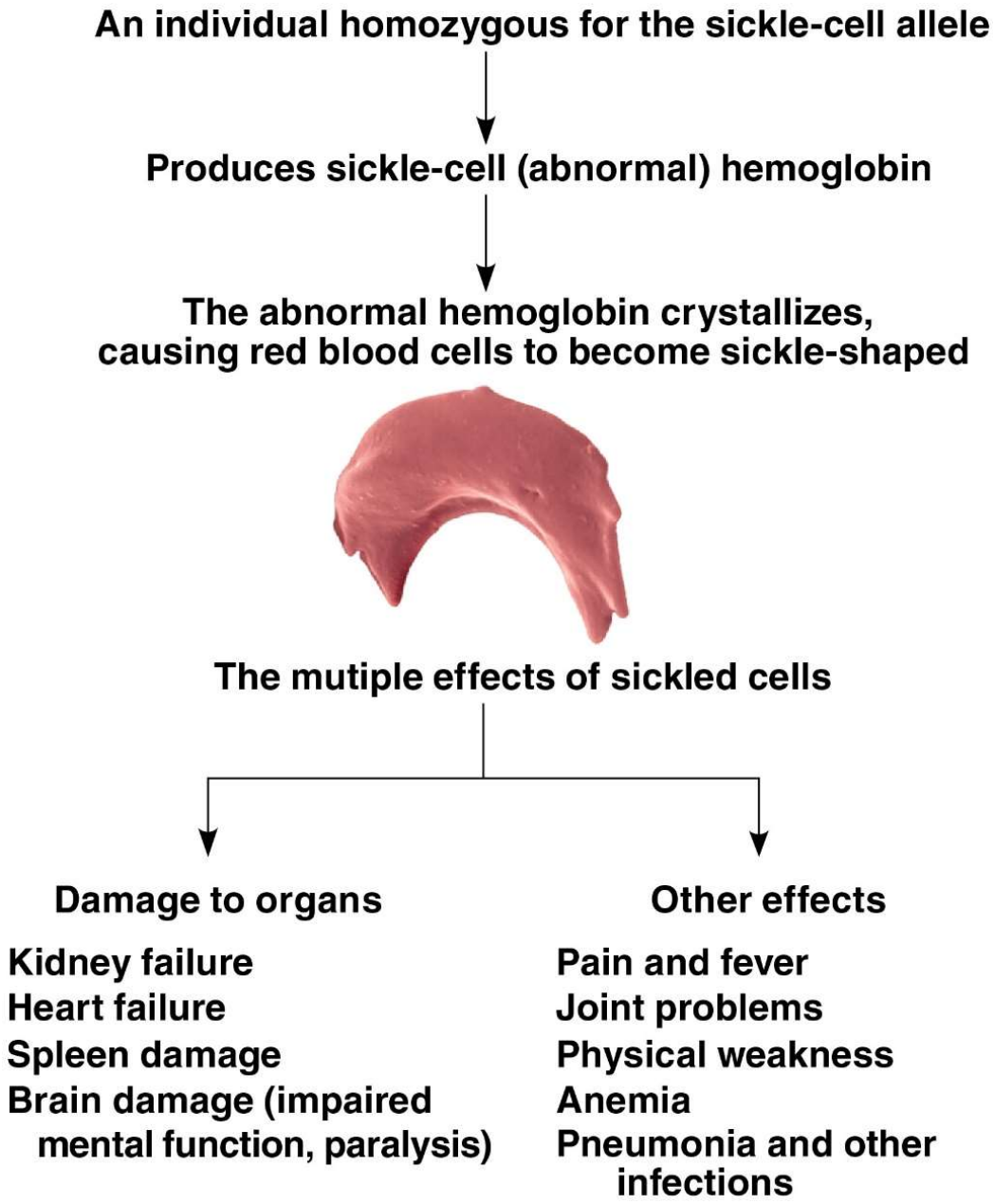
- **Pleiotropy** occurs when one gene influences multiple characters.
- **Sickle-cell disease** is a human example of pleiotropy.
  - This disease affects the type of hemoglobin produced and the shape of red blood cells, and causes anemia and organ damage.
  - Sickle-cell and nonsickle alleles are codominant.
  - Carriers of sickle-cell disease have increased resistance to malaria.

9.13 A Single Gene May Affect Many Phenotypic Characters (2 of 2)

Checkpoint question Why is the sickle-cell trait considered codominant at the molecular level?

Codominance means that both traits are expressed; a carrier for the sickle-cell allele produces both normal and abnormal hemoglobin.

Figure 9.13b



9.14 A Single Character May Be Influenced by Many Genes (1 of 2)

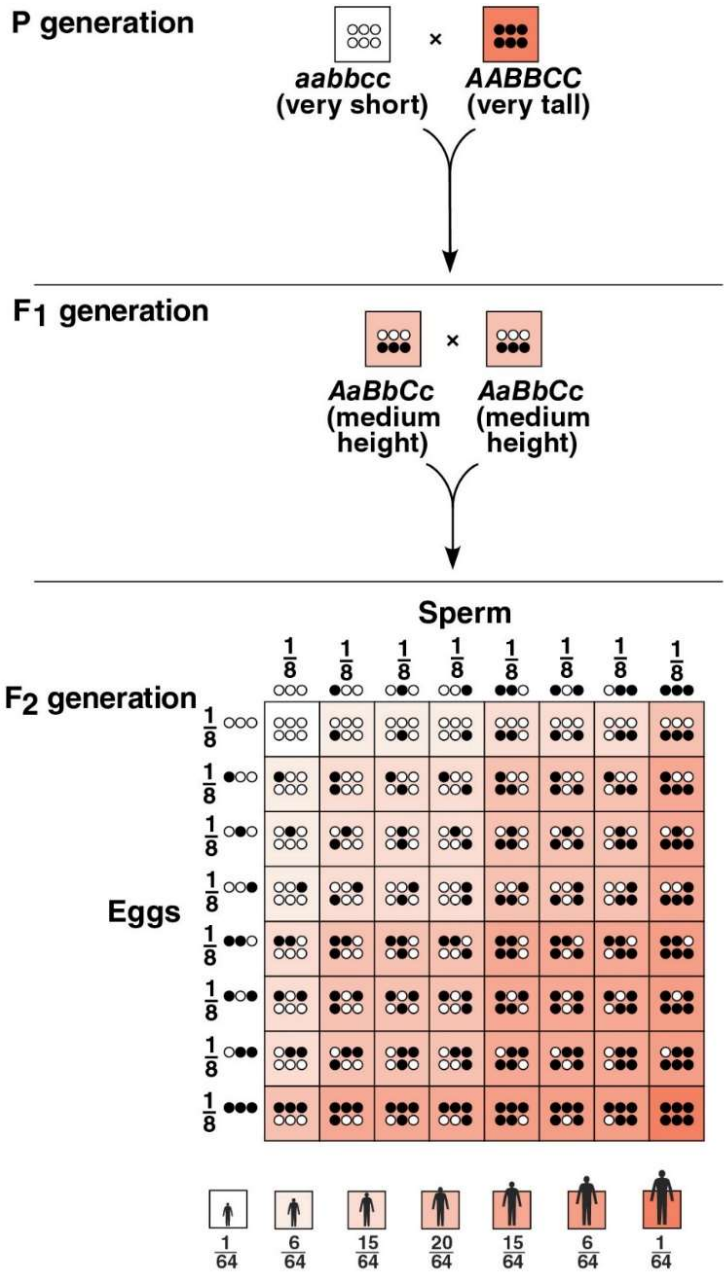
- Many characters result from **polygenic inheritance**, in which a single phenotypic character results from the additive effects of two or more genes on a single phenotypic character.
- Human height is an example of polygenic inheritance.

9.14 A Single Character May Be Influenced by Many Genes (2 of 2)

Checkpoint question An *AaBbcc* individual would be indistinguishable in phenotype from which of the following individuals: *AAbbcc*, *aaBBcc*, *AabbCc*, *Aabbcc*, or *aaBbCc*?

All except *Aabbcc*

Figure 9.14



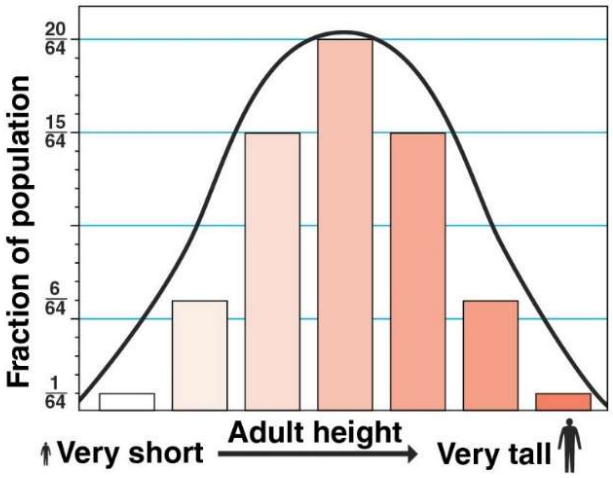
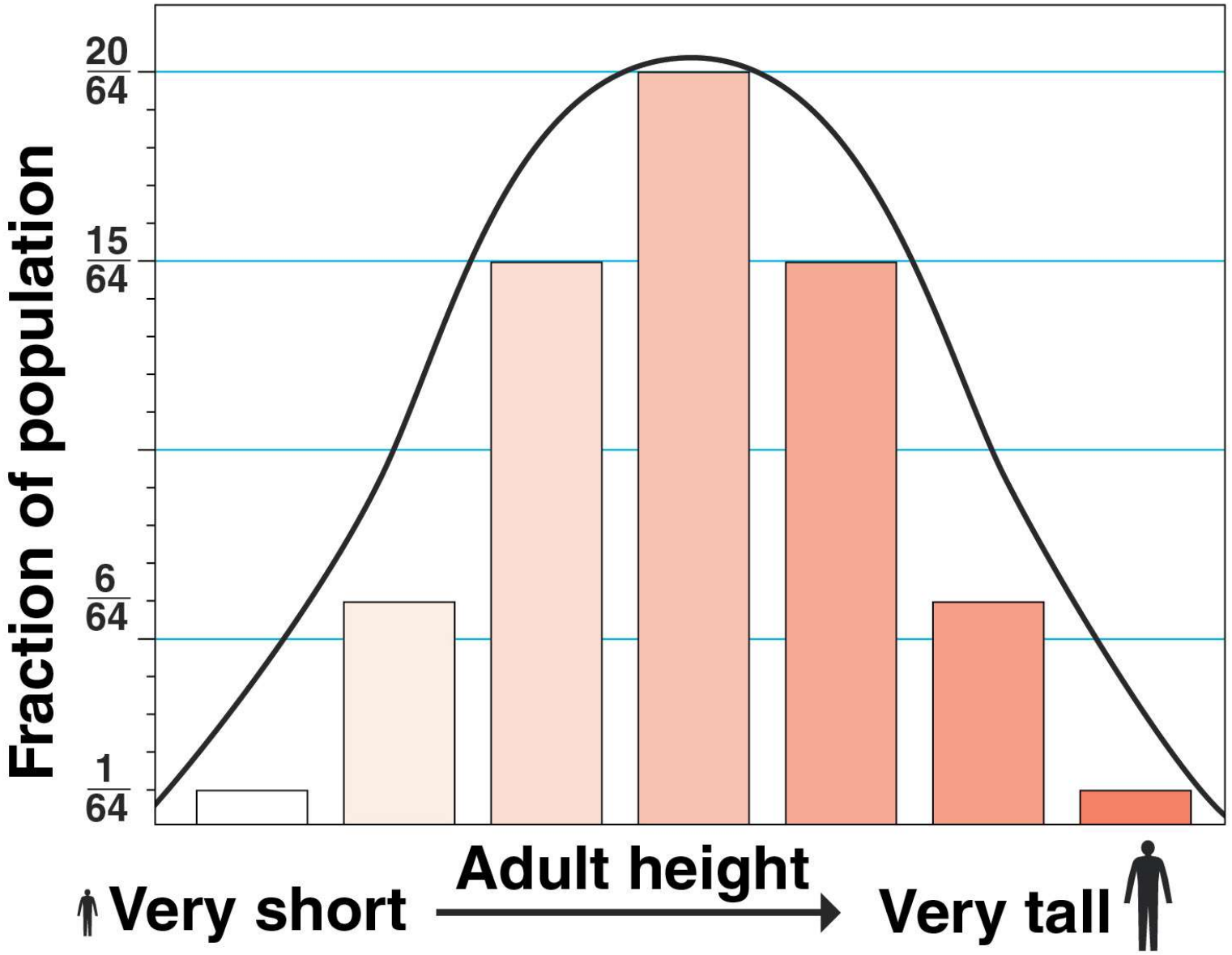


Figure 9.14_3



9.15 The Environment Affects Many Characters

- Many traits are affected, in varying degrees, by both genetic and environmental factors.

Checkpoint question If most characters result from a combination of environment and heredity, why was Mendel able to ignore environmental influences in his pea plants?

The characters he chose for study were all entirely genetically determined and all his test subjects were raised in a similar environment.
Student Misconceptions and Concerns

The Chromosomal Basis of Inheritance

9.16 Chromosome Behavior Accounts for Mendel's Laws (1 of 2)

- The **chromosome theory of inheritance** holds that
  - genes occupy specific loci (positions) on chromosomes and
  - chromosomes undergo segregation and independent assortment during meiosis.
- Mendel's laws correlate with chromosome separation in meiosis.

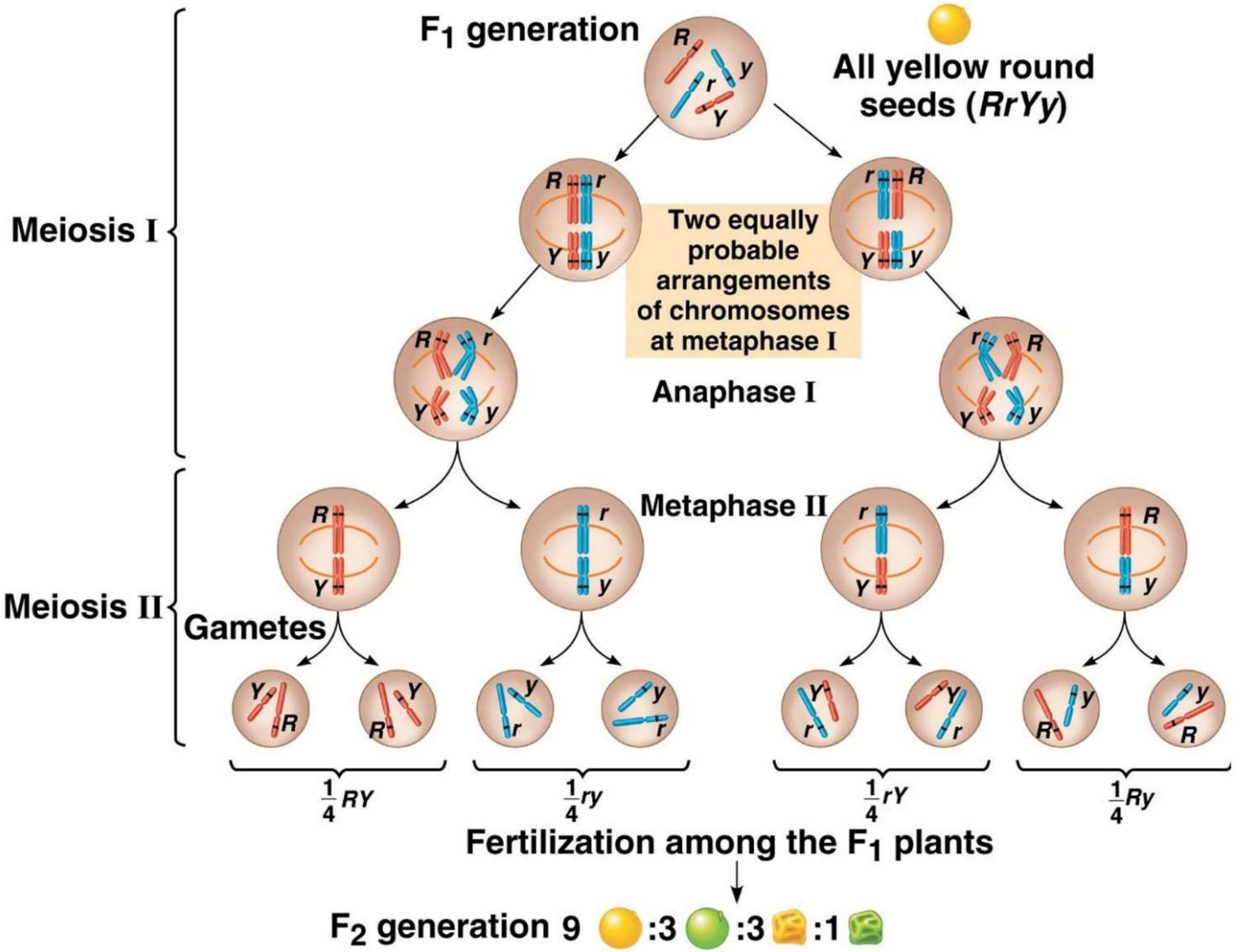
9.16 Chromosome Behavior Accounts for Mendel's Laws (2 of 2)

Checkpoint question Which of Mendel's laws have their physical basis in the following phases of meiosis:

a. the orientation of homologous chromosome pairs in metaphase I;
b. the separation of homologous chromosomes in anaphase I?

(a) The law of independent assortment; (b) the law of segregation

Figure 9.16_1_3



9.17 Scientific Thinking: Genes on the same Chromosome Tend to Be Inherited Together (1 of 2)

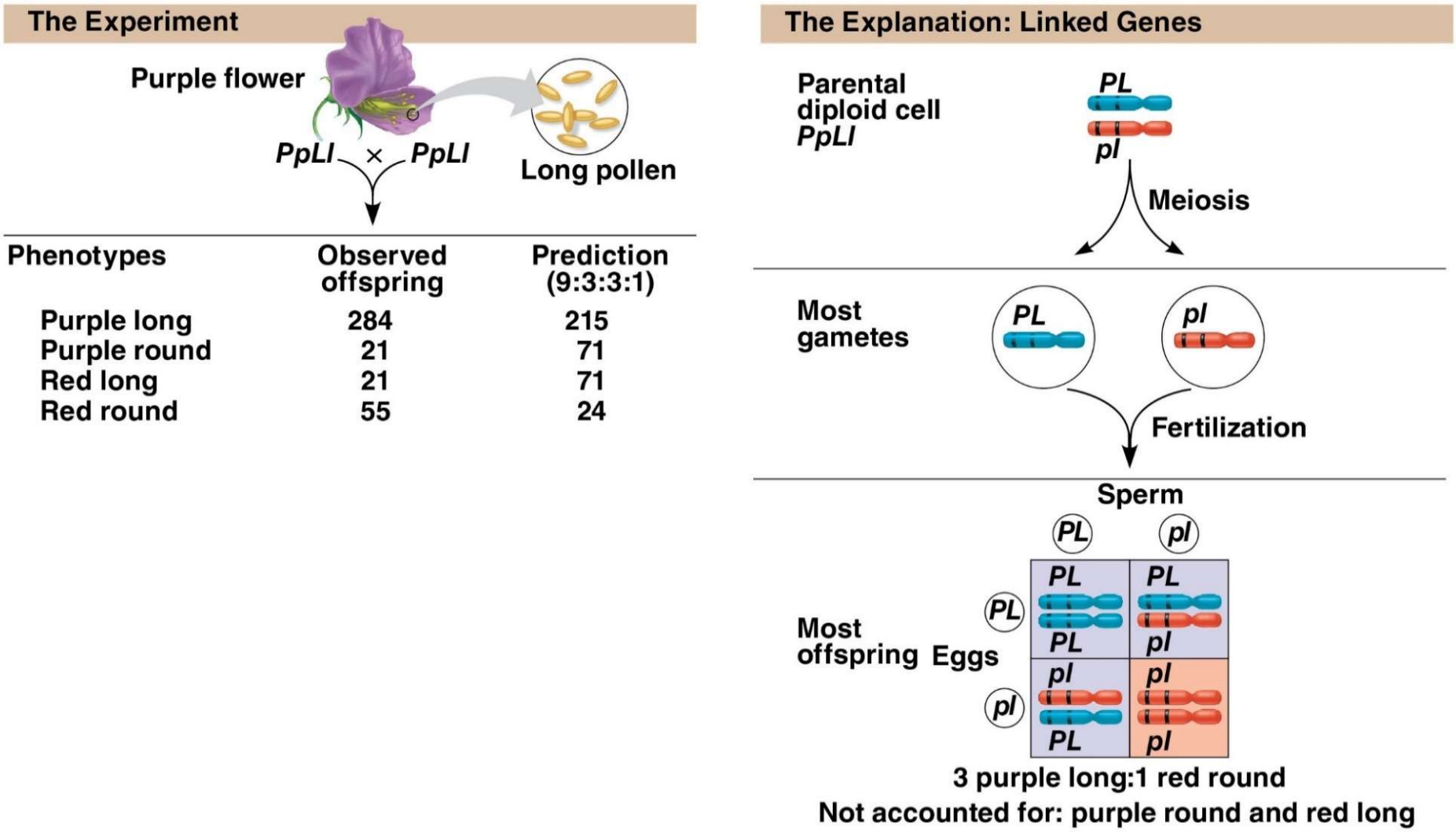
- Bateson and Punnett studied plants that did not show a 9:3:3:1 ratio in the F_2 generation. What they found was an example of **linked genes**, which
  - are located close together on the same chromosome and
  - tend to be inherited together.

9.17 Scientific Thinking: Genes on the same Chromosome Tend to Be Inherited Together (2 of 2)

Checkpoint question In what way was Bateson and Punnett's success dependent upon failing at first?

The "failure" to obtain the expected results provided the insight that led to the discovery of linked genes.

Figure 9.17



9.18 Crossing Over Produces New Combinations of Alleles (1 of 2)

- Crossing over between homologous chromosomes produces new combinations of alleles in gametes.
- Linked genes can be separated by crossing over, forming recombinant gametes.
- The percentage of recombinant offspring among the total is the **recombination frequency**.

Figure 9.18a

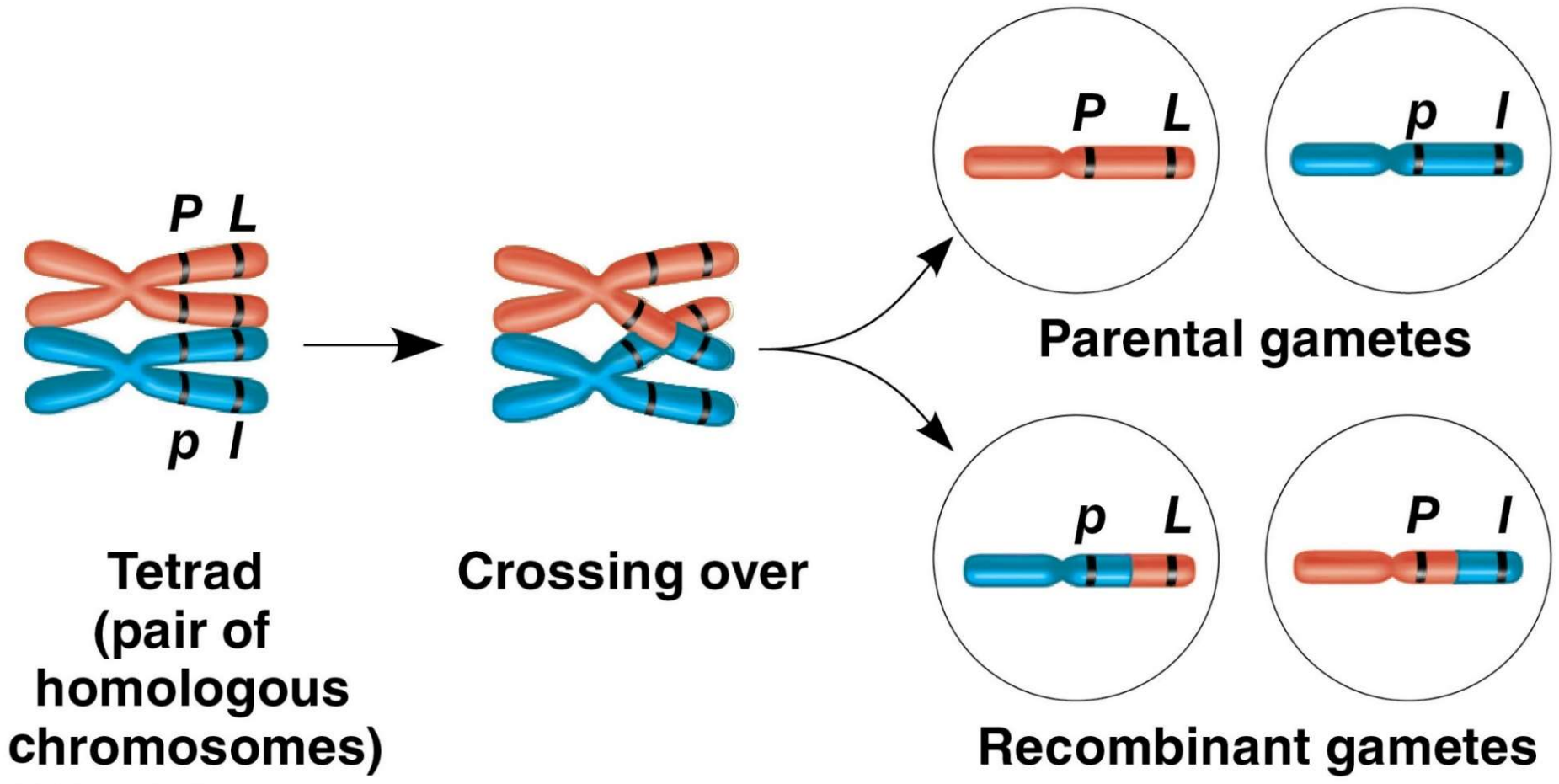
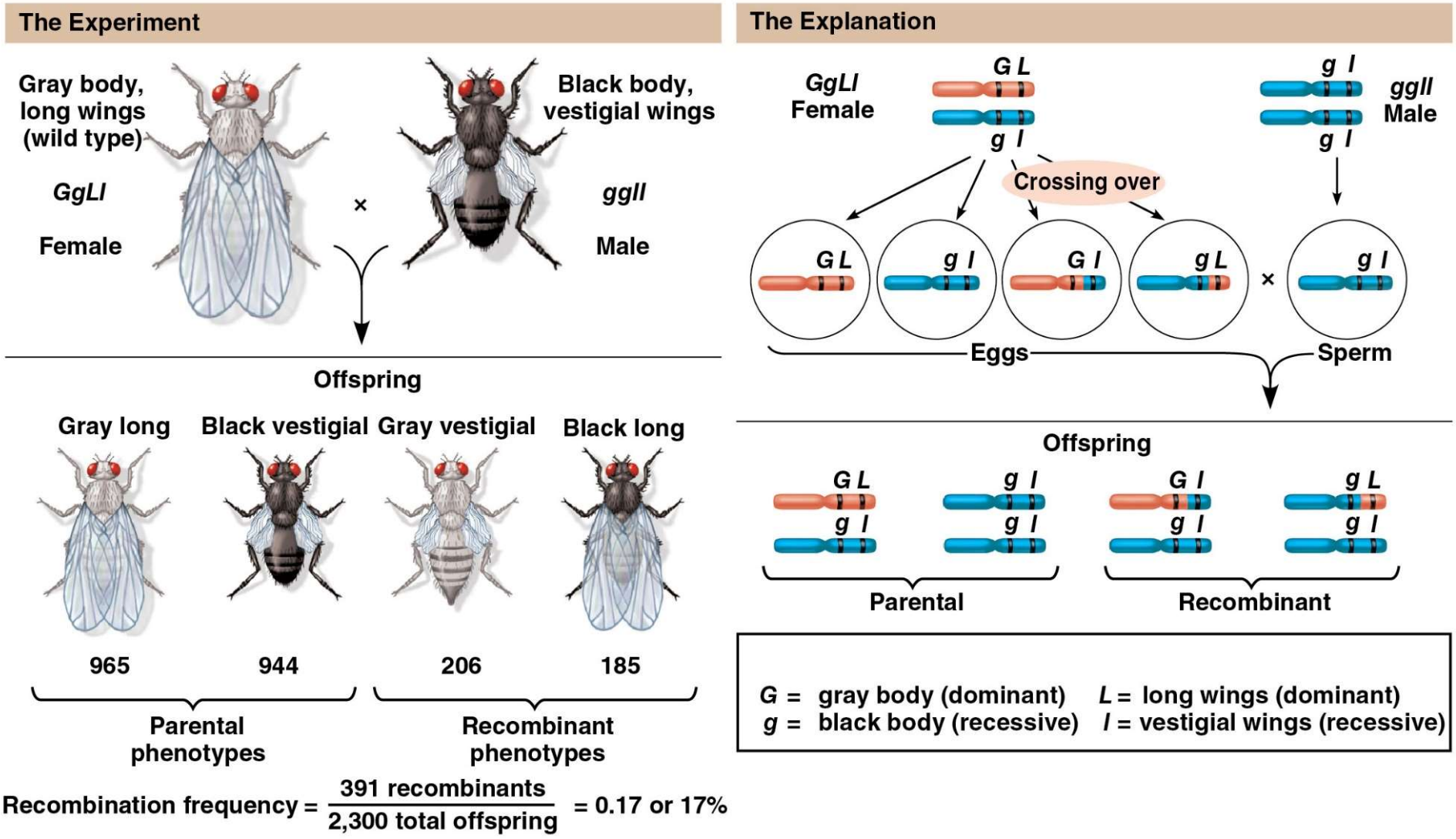
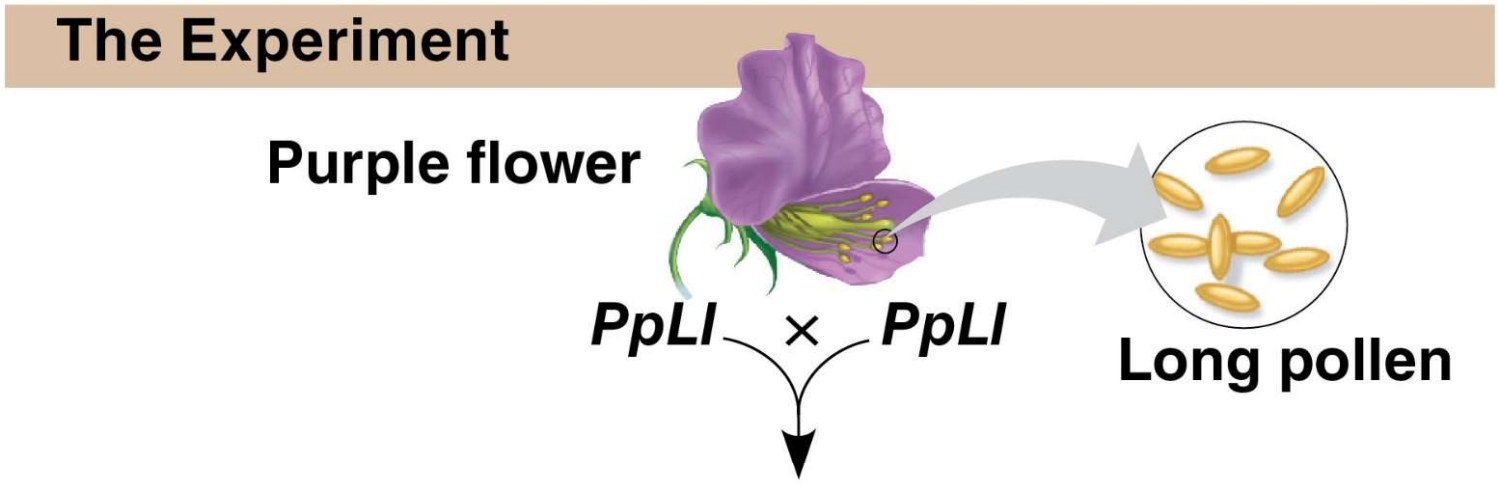


Figure 9.18c



9.18 Crossing Over Produces New Combinations of Alleles (2 of 2)



Phenotypes	Observed offspring	Prediction (9:3:3:1)
Purple long	284	215
Purple round	21	71
Red long	21	71
Red round	55	24

Checkpoint question Return to the data in Figure 9.17. What is the recombination frequency between the flower-color and pollen-length genes?

9.19 Geneticists Use Crossover Data to Map Genes (1 of 2)

- Recombination frequencies can be used to map the relative positions of genes on chromosomes.
  - A **genetic map** is an ordered list of the genetic loci along a chromosome.
  - Such a genetic map based on recombinant frequencies is called a **linkage map**.

Figure 9.19a

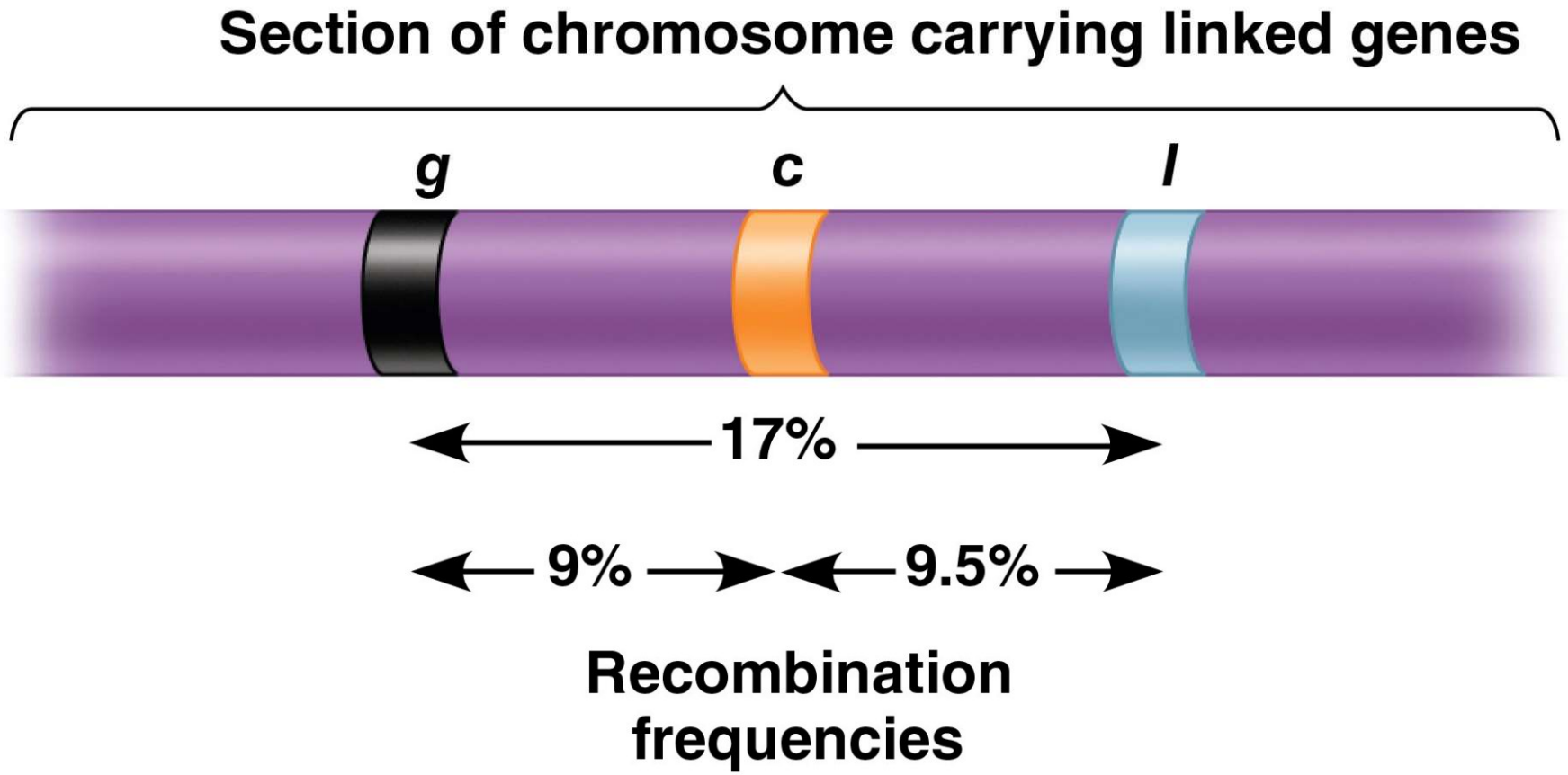
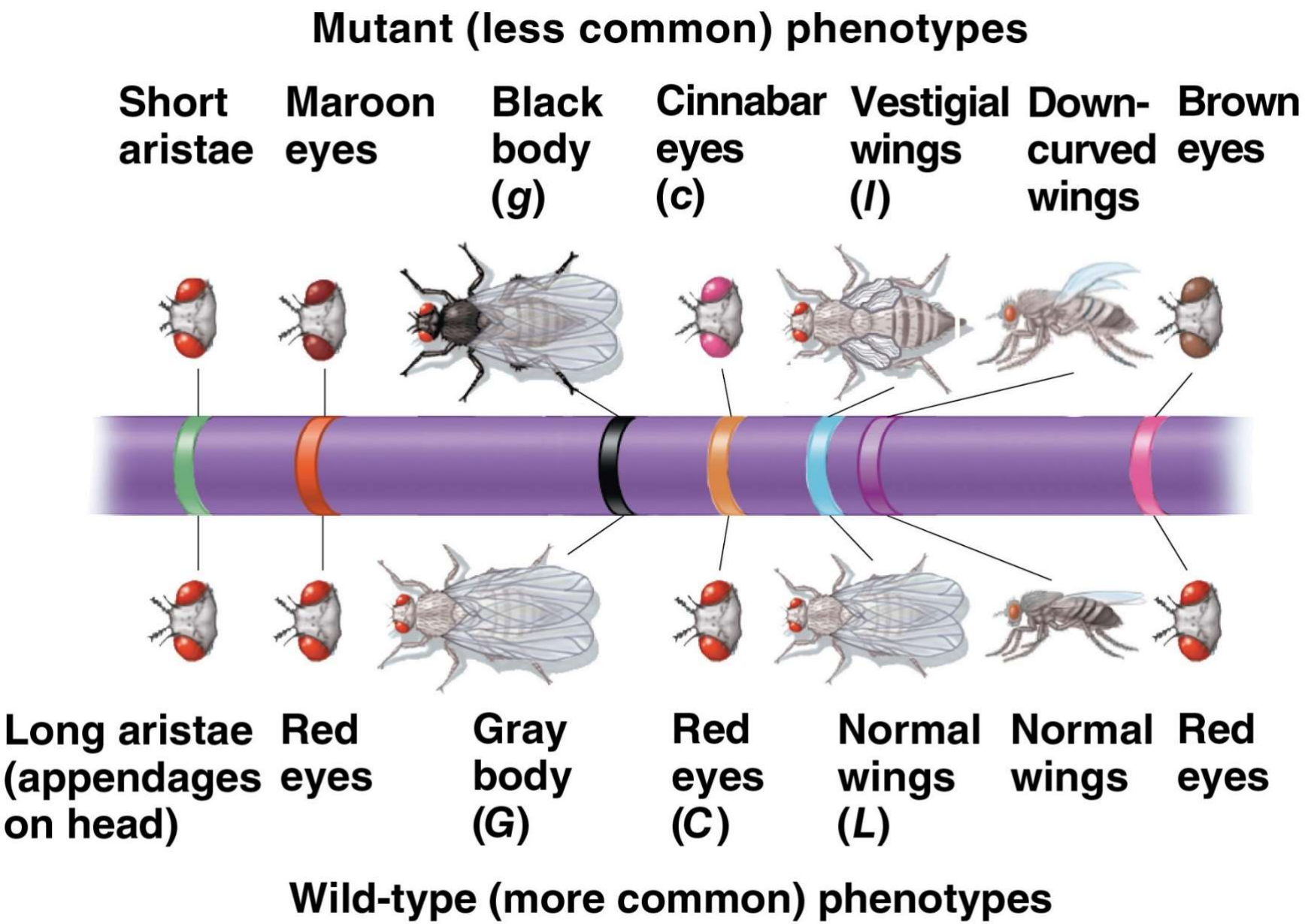
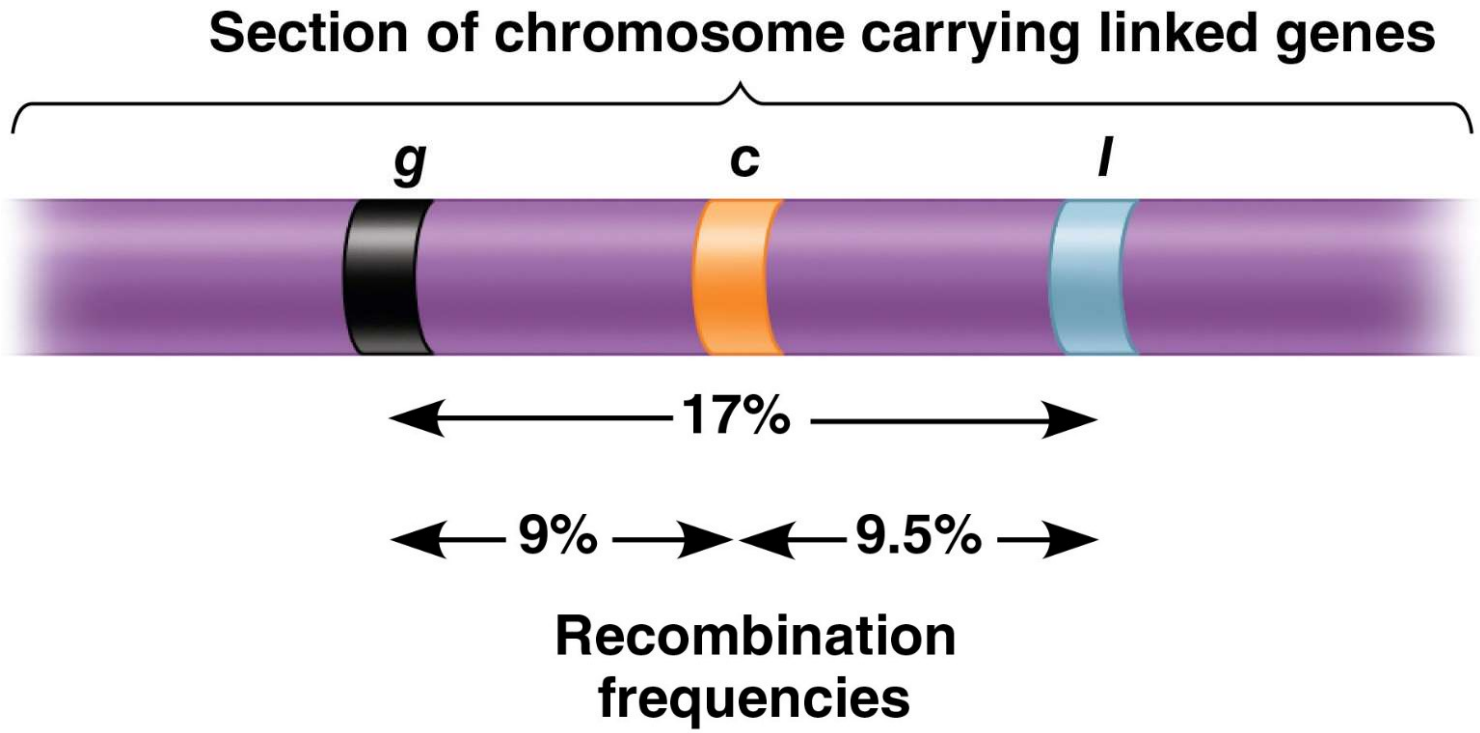


Figure 9.19b



9.19 Geneticists Use Crossover Data to Map Genes (2 of 2)



Checkpoint question You design *Drosophila* crosses to provide recombination data for a gene not included in Figure 9.19A. The gene has recombination frequencies of 3% with the vestigial-wing (*l*) locus and 7% with the cinnabar-eye (*c*) locus. Where is it located on the chromosome?

Sex Chromosomes and Sex-Linked Genes

9.20 Chromosomes Determine Sex in Many Species (1 of 3)

- In mammals, a male has XY **sex chromosomes**, and a female has XX.
  - The Y chromosome has genes for the development of testes, whereas an absence of the Y allows ovaries to develop.
  - In addition, human males and females both have 44 autosomes (nonsex chromosomes).
- Many non-mammalian species have other chromosomal systems that determine sex.

9.20 Chromosomes Determine Sex in Many Species (2 of 3)

- In some animals, environmental temperature determines the sex.
  - For some reptiles, the temperature at which eggs are incubated during a specific period of embryonic development determines whether that embryo will develop into a male or female.
  - Global climate change may therefore impact the sex ratio of such species.

9.20 Chromosomes Determine Sex in Many Species (3 of 3)

Checkpoint question King Henry VIII of England was quick to blame his wives for bearing him only daughters. Explain how, from a genetic point of view, his thinking was wrong.

9.21 Sex-linked Genes Exhibit a Unique Pattern of Inheritance (1 of 2)

- A gene located on either sex chromosome is called a **sex-linked gene**.
- The X chromosome carries many **X-linked genes** that control traits unrelated to sex.
- The inheritance of white eye color in the fruit fly illustrates an X-linked recessive trait.

9.21 Sex-linked Genes Exhibit a Unique Pattern of Inheritance (2 of 2)

Checkpoint question A white-eyed female *Drosophila* is mated with a red-eyed (wild-type) male. What result do you predict for the numerous offspring?

All female offspring will be red-eyed but heterozygous ($X^R X^r$); all male offspring will be white-eyed ($X^r Y$).

9.22 Connection: Human Sex-linked Disorders Affect Mostly Males (1 of 2)

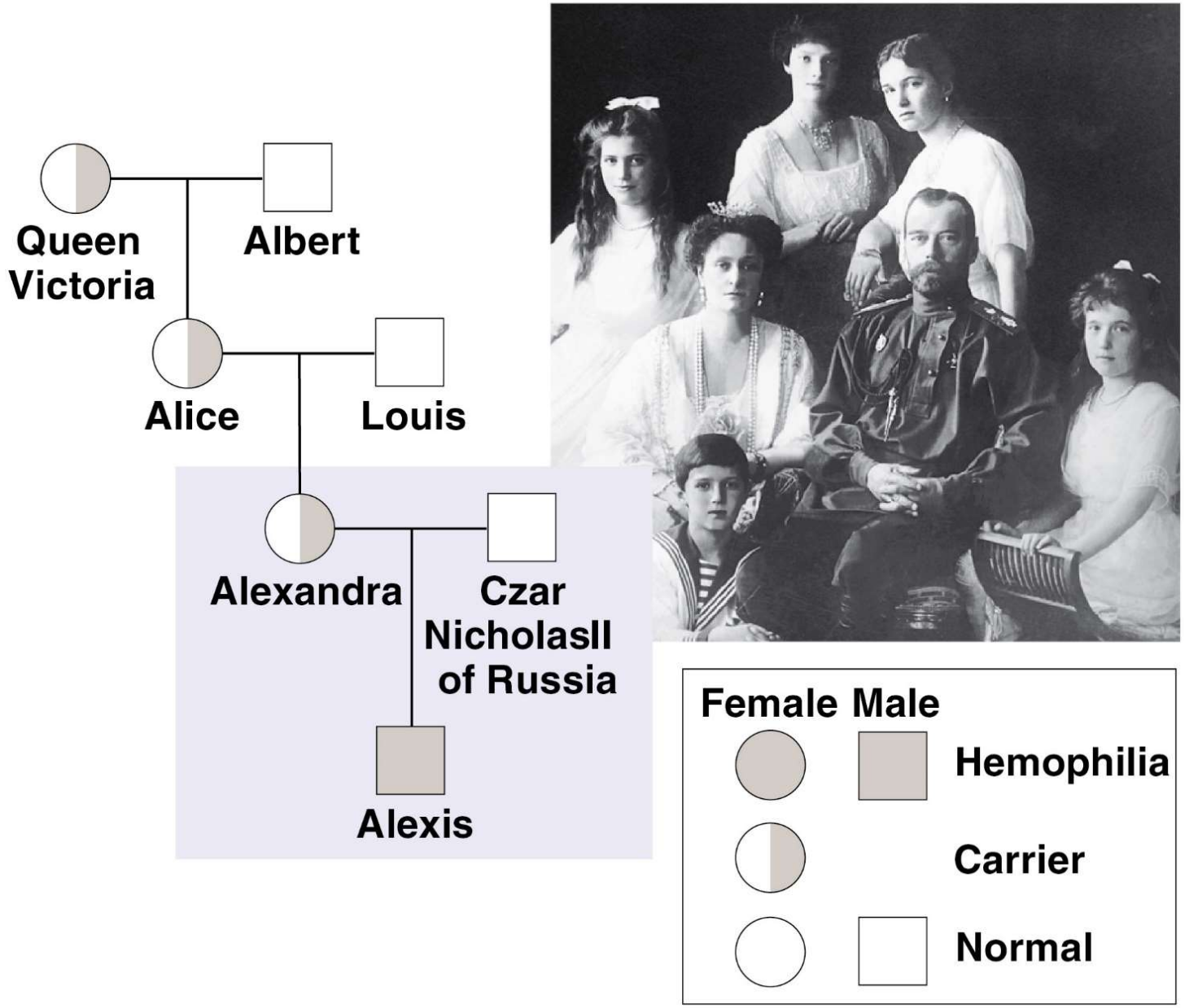
- Most X-linked human disorders are due to recessive alleles and therefore are seen mostly in males.
  - A male receiving a single X-linked recessive allele from his mother will have the disorder.
  - A female must receive the allele from both parents to be affected.

9.22 Connection: Human Sex-linked Disorders Affect Mostly Males (2 of 2)

Checkpoint question Neither Tom nor Sue has hemophilia, but their first son does. If the couple has a second child, what is the probability that he or she will also have the disease?

¼ (½ chance of a male child × ½ chance that he will inherit the mutant X)

Figure 9.22



9.23 Evolution Connection: The Y Chromosome Provides Clues about Human Male Evolution (1 of 2)

- Y chromosomes can provide data about recent human evolutionary history because they are passed on intact from father to son.

9.23 Evolution Connection: The Y Chromosome Provides Clues about Human Male Evolution (2 of 2)

- In 2003, geneticists discovered that about 8% of males currently living in central Asia have Y chromosomes of striking genetic similarity.
  - Further analysis traced their common genetic heritage to a man living about 1,000 years ago.
  - In combination with historical records, the data led to the speculation that the Mongolian ruler Genghis Khan may be responsible for the spread of the telltale chromosome to nearly 16 million male descendants.