Chapter 14

Mendel and the Gene Idea

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PowerPoint® Lecture Presentations for

Biology

Eighth Edition

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Concept 14.1: Mendel used the scientific approach to identify two laws of inheritance

- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments (mid 1800's)

- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**
- The true-breeding parents are the **P generation**
- The hybrid offspring of the P generation are called the **F_1 generation**
- When F_1 individuals self-pollinate, the **F_2 generation** is produced (in animals, F1 x F1 means a brother/sister mating!)

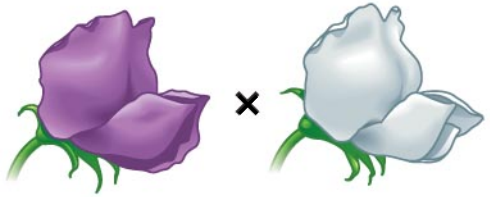
The Law of Segregation

- When Mendel crossed contrasting, true-breeding white and purple flowered pea plants, all of the F_1 hybrids were purple
- When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had white
- Mendel discovered a ratio of about three to one, purple to white flowers, in the F_2 generation

Fig. 14-3-1

EXPERIMENT

P Generation (true-breeding parents)



Purple flowers × **White flowers**

Fig. 14-3-2

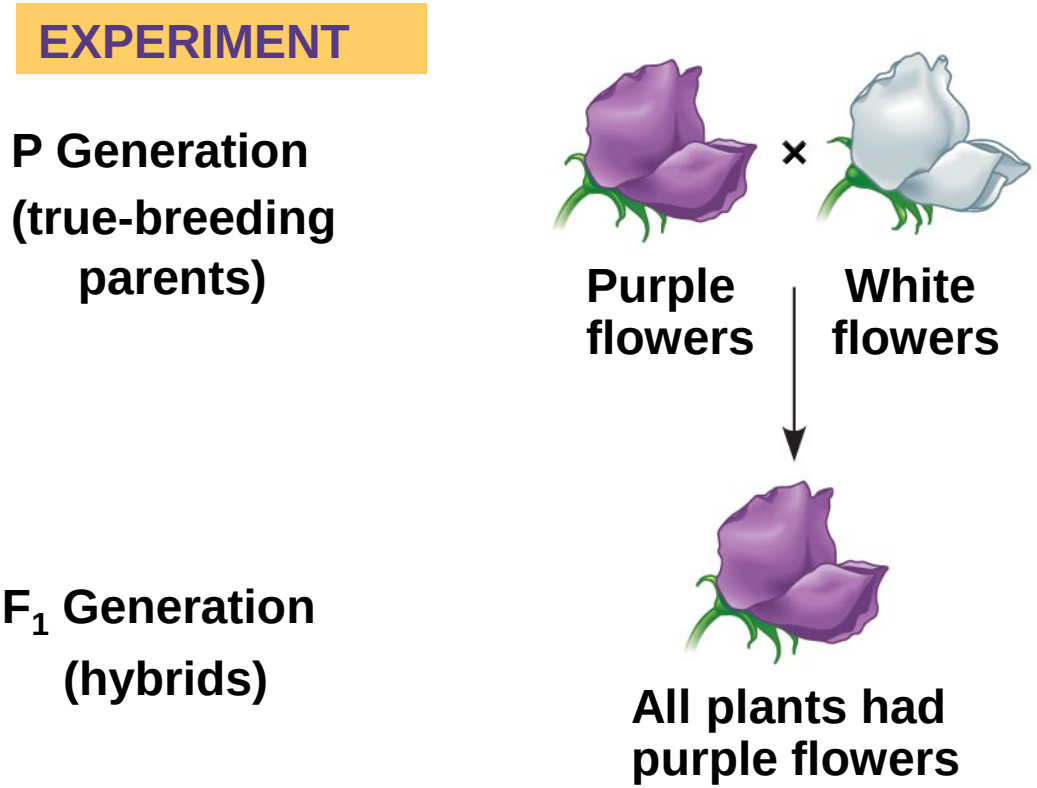
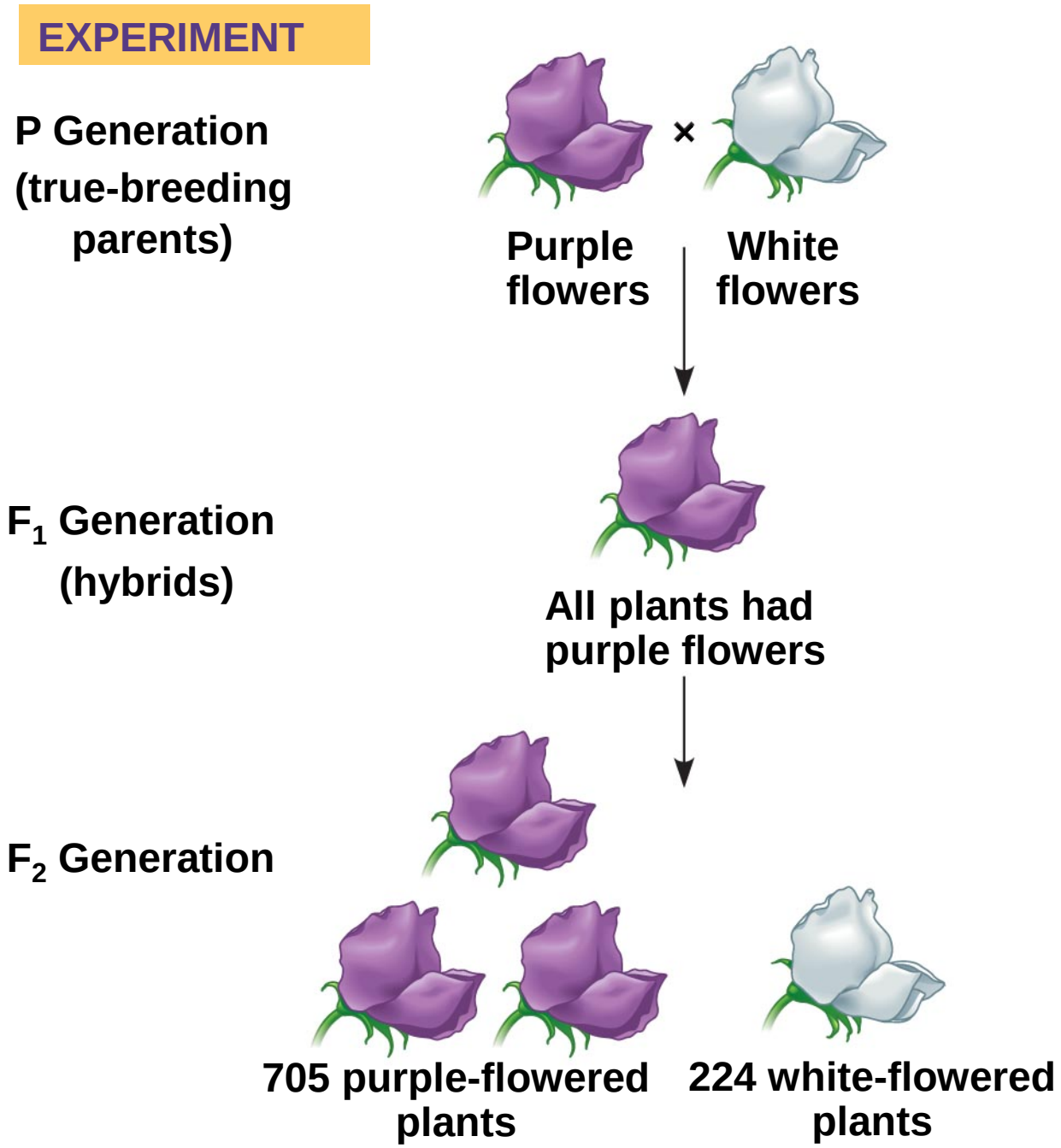


Fig. 14-3-3



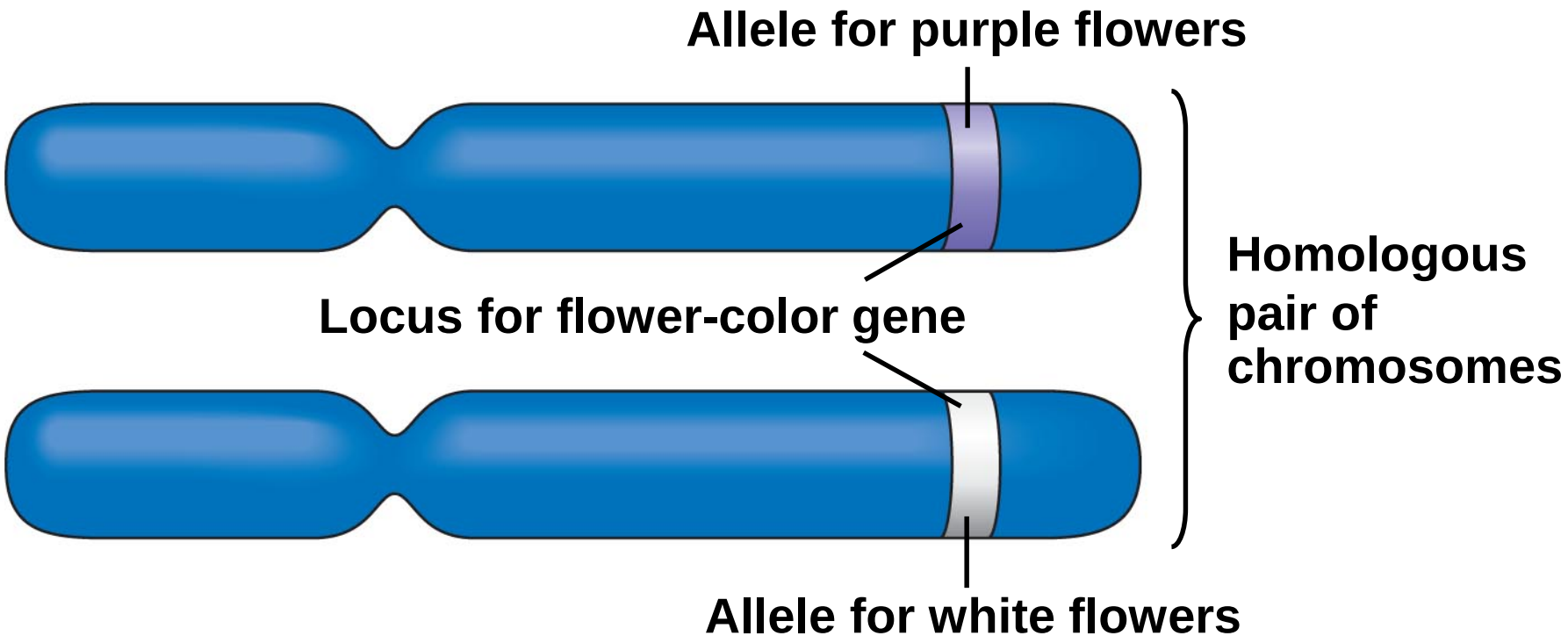
- Mendel reasoned that only the purple flower factor was affecting flower color in the F_1 hybrids
- Mendel called the purple flower color a dominant trait and the white flower color a recessive trait
- What Mendel called a “heritable factor” is what we now call a gene

Mendel's Model

- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F_2 offspring
- Four related concepts make up this model

- The first concept is that alternative versions of genes account for variations in inherited characters
- For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers
- These alternative versions of a gene are now called **alleles**
- Each gene resides at a specific locus on a specific chromosome

Fig. 14-4



- The second concept is that for each character an organism inherits two alleles, one from each parent
- Mendel made this deduction without knowing about the role of chromosomes
- The two alleles at a locus on a chromosome may be identical, as in the true-breeding plants of Mendel's P generation
- Alternatively, the two alleles at a locus may differ, as in the F_1 hybrids

- The third concept is that if the two alleles at a locus differ, then one (the **dominant allele**) determines the organism's appearance, and the other (the **recessive allele**) has no noticeable effect on appearance
- In the flower-color example, the F_1 plants had purple flowers because the allele for that trait is dominant

- The fourth concept, now known as the **law of segregation**, states that the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes
- Thus, an egg or a sperm gets only one of the two alleles that are present in the somatic cells of an organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

- Mendel's segregation model accounts for the 3:1 ratio he observed in the F_2 generation of his numerous crosses
- The possible combinations of sperm and egg can be shown using a **Punnett square**, a diagram for predicting the results of a genetic cross between individuals of known genetic makeup
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele

Fig. 14-5-1

P Generation

Appearance: Purple flowers × White flowers

Genetic makeup: *PP* *pp*

Gametes: *P* *p*

Fig. 14-5-2

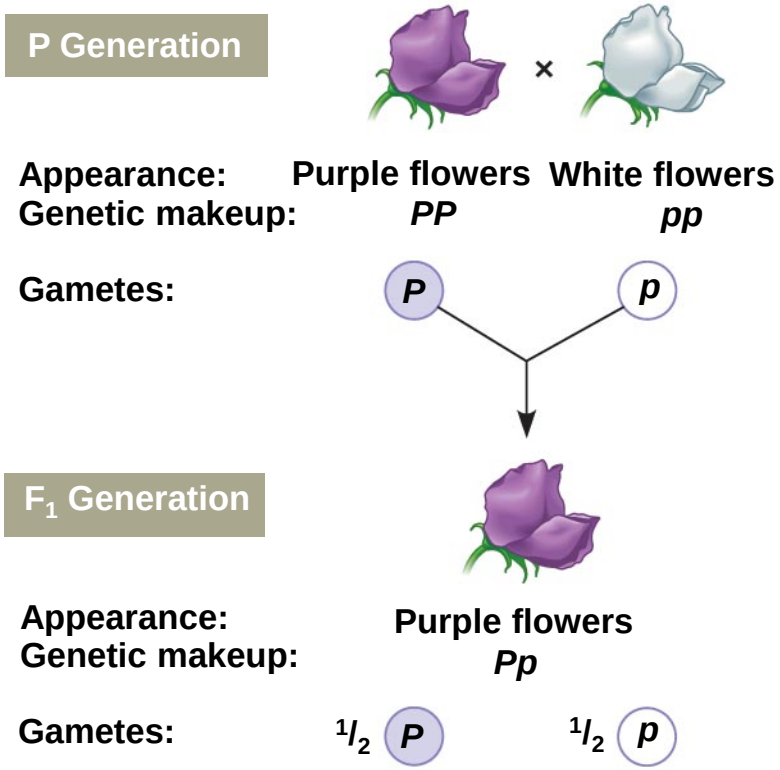
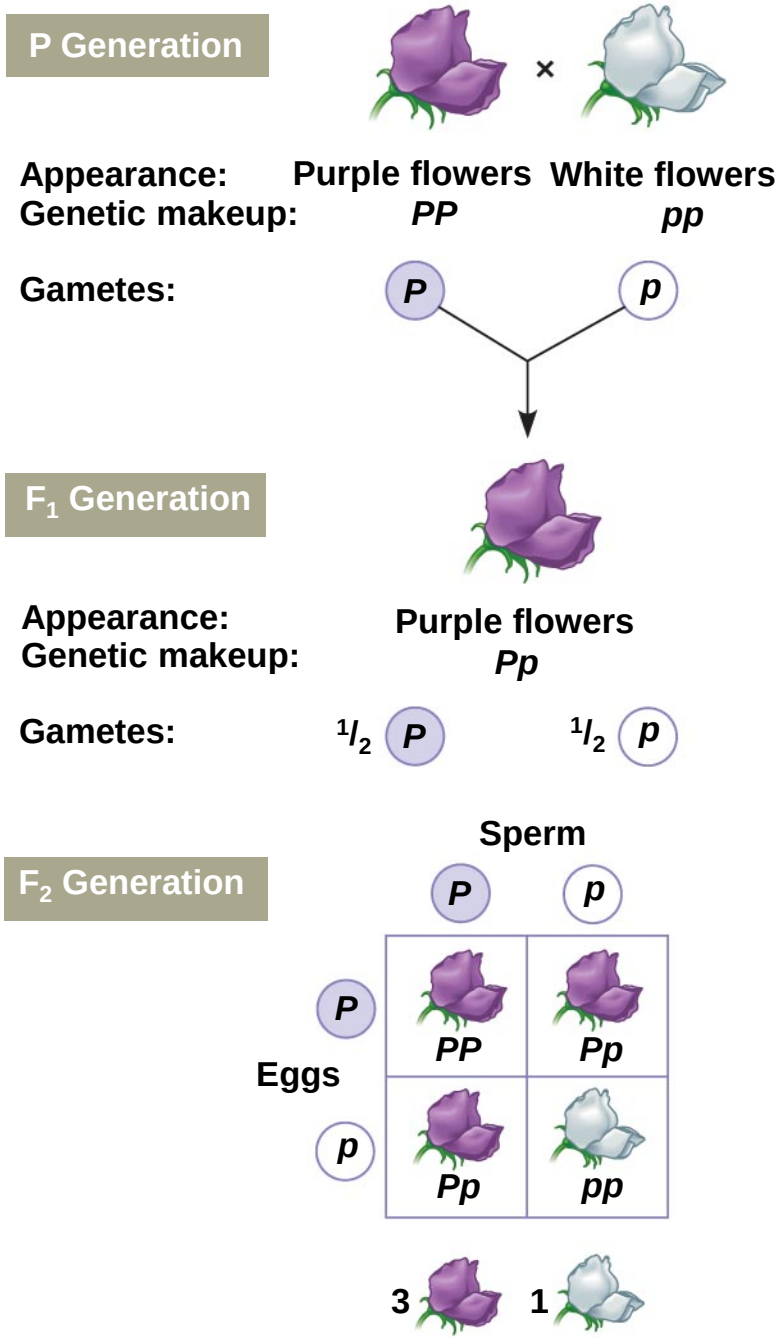


Fig. 14-5-3

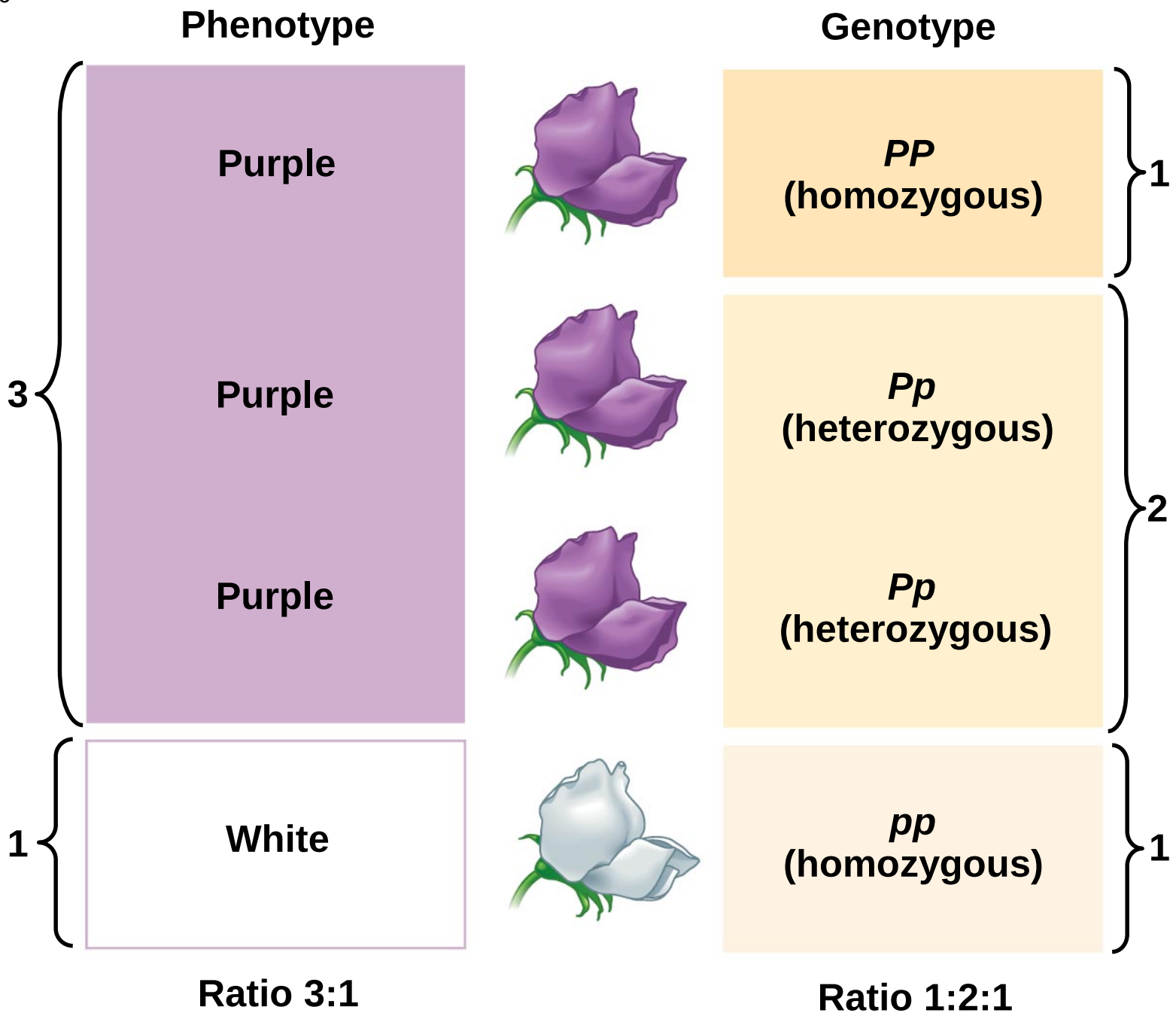


Useful Genetic Vocabulary

- An organism with two identical alleles for a character is said to be **homozygous** for the gene controlling that character
- An organism that has two different alleles for a gene is said to be **heterozygous** for the gene controlling that character
- Unlike homozygotes, heterozygotes are not true-breeding

- Because of the different effects of dominant and recessive alleles, an organism's traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism's **phenotype**, or physical appearance, and its **genotype**, or genetic makeup
- In the example of flower color in pea plants, *PP* and *Pp* plants have the same phenotype (purple) but different genotypes

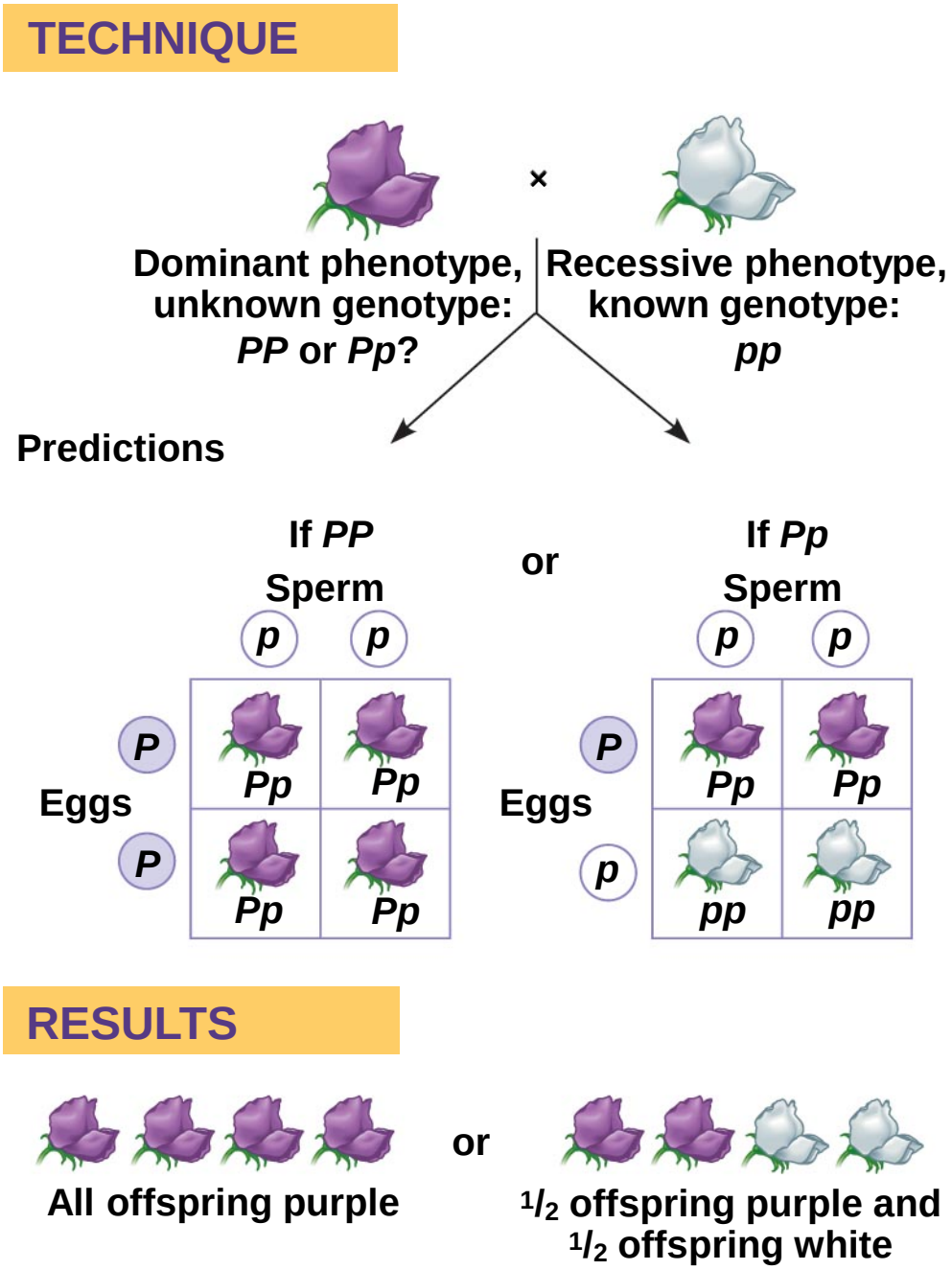
Fig. 14-6



The Testcross

- How can we tell the genotype of an individual with the dominant phenotype?
- Such an individual must have one dominant allele, but the individual could be either homozygous dominant or heterozygous
- The answer is to carry out a **testcross**: breeding the mystery individual with a homozygous recessive individual
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous

Fig. 14-7



The Law of Independent Assortment

- Mendel derived the law of segregation by following a single character
- The F_1 offspring produced in this cross were **monohybrids**, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a *monohybrid cross*

- Mendel identified his second law of inheritance by following two characters at the same time
- Crossing two true-breeding parents differing in two characters produces **dihybrids** in the F_1 generation, heterozygous for both characters
- A dihybrid cross, a cross between F_1 dihybrids, can determine whether two characters are transmitted to offspring as a package or independently

Fig. 14-8

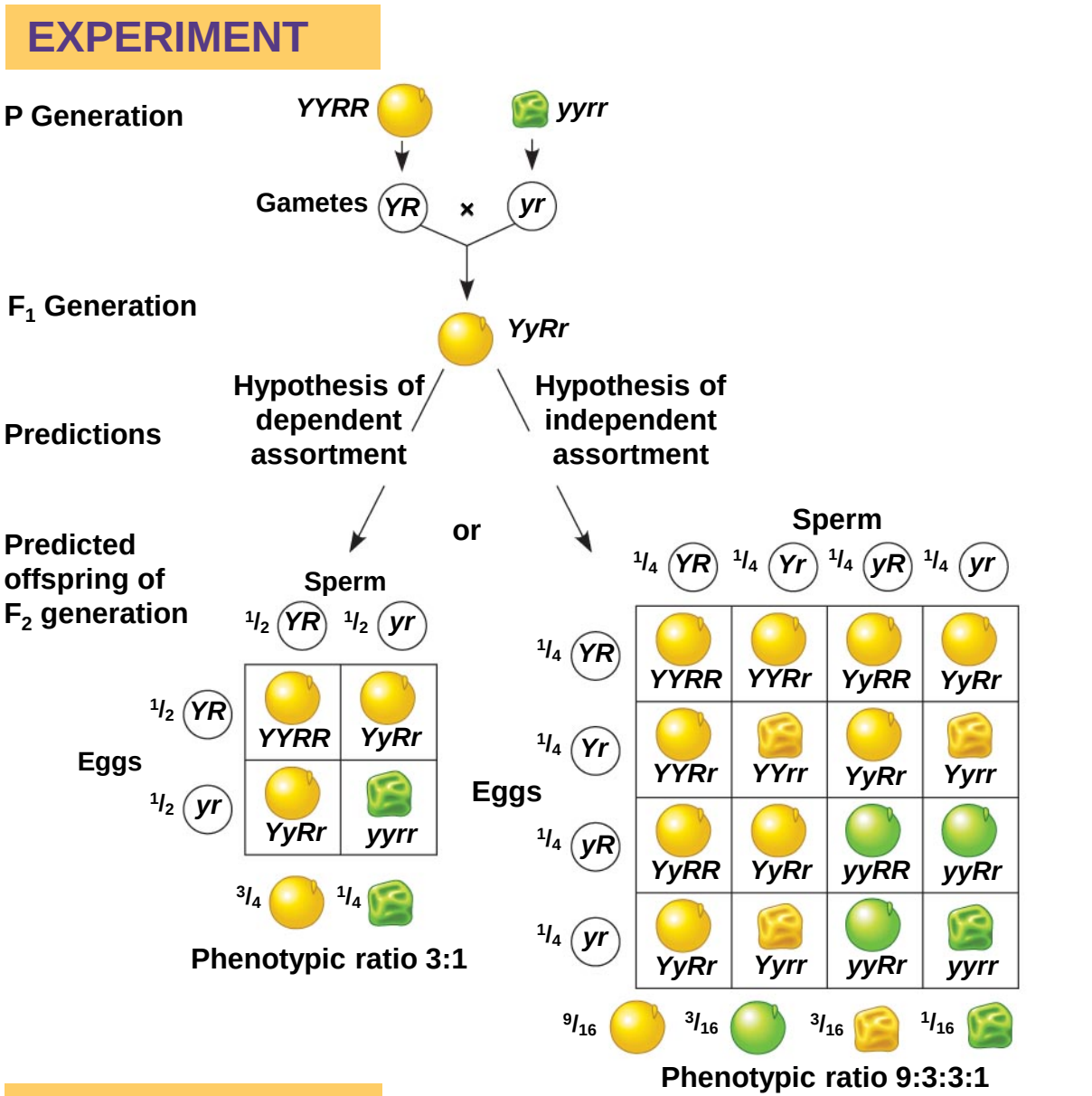


Fig. 14-8a

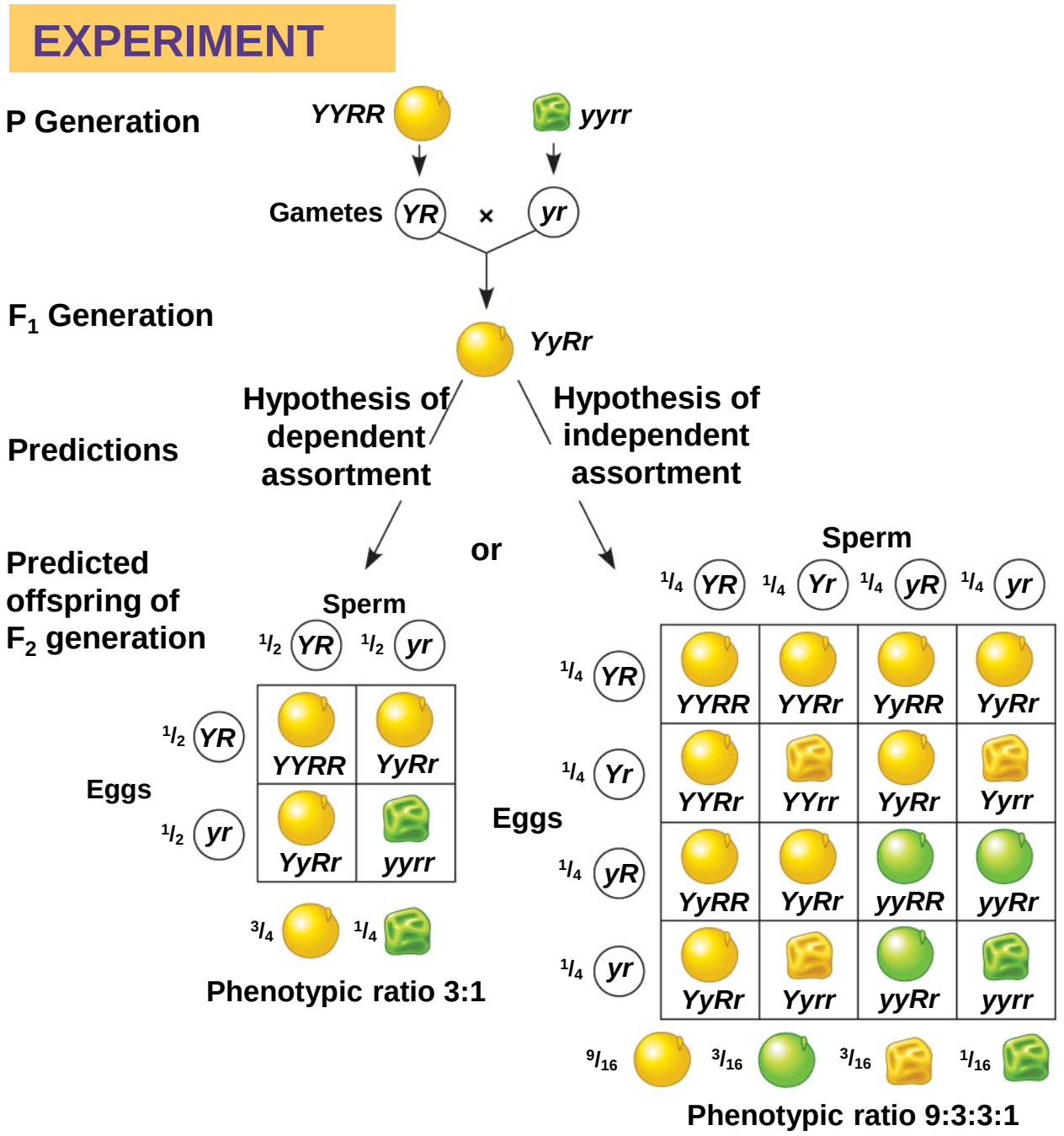


Fig. 14-8b

RESULTS

315  108  101  32 

Phenotypic ratio approximately 9:3:3:1

- Using a dihybrid cross, Mendel developed the **law of independent assortment**
- The law of independent assortment states that each pair of alleles segregates independently of each other pair of alleles during gamete formation
- Strictly speaking, this law applies only to genes on different, nonhomologous chromosomes
- Genes located near each other on the same chromosome tend to be inherited together (linkage)

Concept 14.2: The laws of probability govern Mendelian inheritance

- Mendel's laws of segregation and independent assortment reflect the rules of probability
- When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss
- In the same way, the alleles of one gene segregate into gametes independently of another gene's alleles

Probability

- P (probability) = $\frac{\text{\# successful outcomes}}{\text{\# possible outcomes}}$

\# successful outcomes is the number of different ways a specific outcome can occur

\# possible outcomes is the number of all possible outcomes that can occur

$P \leq 1$

Example: a deck of cards has 52 cards

There are 13 of each “suit” (hearts, clubs, spades, diamonds).

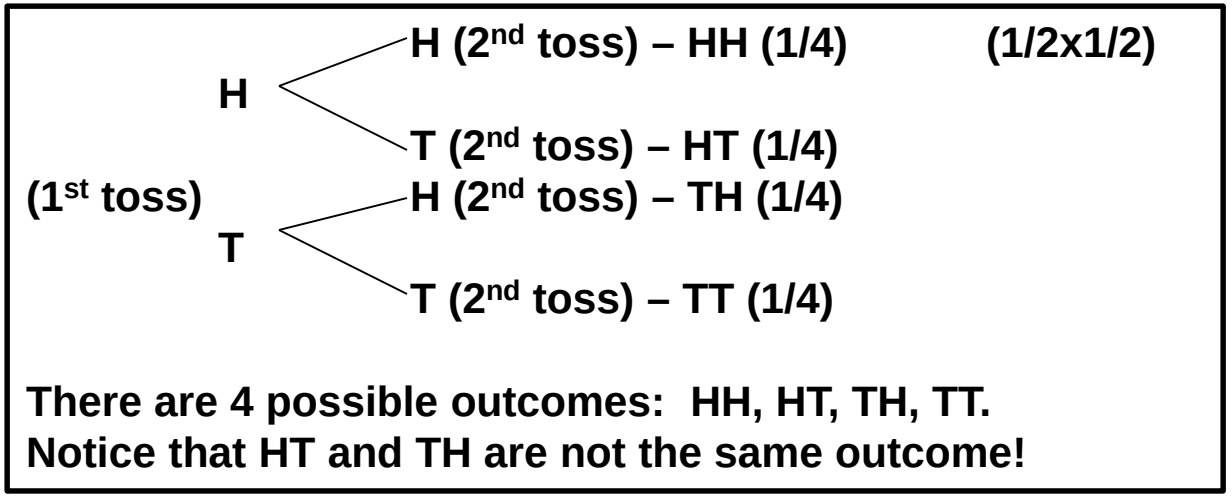
What is the probability of drawing a club? A club and a diamond?

The Multiplication and Addition Rules Applied to Monohybrid Crosses

- The multiplication rule (product rule) states that the probability that two or more independent events will occur together is the product of their individual probabilities
- A fork-line diagram can be used to illustrate.
- Probability in an F_1 monohybrid cross can be determined using the multiplication rule
- Segregation in a heterozygous plant is like flipping a coin: Each gamete has a ½ chance of carrying the dominant allele and a ½ chance of carrying the recessive allele

Tossing a coin

- What is the probability of getting two “heads” in a row when you toss two coins simultaneously or consecutively? Each toss or coin is independent.
- Two “tails”? A “heads” and a “tails”?
- Notice that, in this case, the order of the outcomes matters.



- The rule of addition (sum rule) states that the probability that any one of two or more exclusive events will occur is calculated by adding together their individual probabilities
- If there are two or more pathways to an outcome, the probability of that outcome is equal to the sum of the probabilities of the pathways to it.
- This can be done with a contingency table (aka Punnett square)

Tossing a coin

- What is the probability of getting two “heads” when you toss one coin twice?
- Two “tails”? A “heads” and a “tails”?
- Notice that, in this case, the order of the outcomes does not matter. HT and TH are the same (as with alleles). Therefore this probability (H+T) is ½.

		2nd toss: H (1/2)	2nd toss: T (1/2)
1st toss	H (1/2)	HH (1/4)	HT (1/4)
	T (1/2)	TH (1/4)	TT (1/4)

Solving Complex Genetics Problems with the Rules of Probability

- We can apply the multiplication and addition rules to predict the outcome of crosses involving multiple characters
- A dihybrid or other multicharacter cross is equivalent to two or more independent monohybrid crosses occurring simultaneously
- In calculating the chances for various genotypes, each character is considered separately, and then the individual probabilities are multiplied together

Concept 14.3: Inheritance patterns are often more complex than predicted by simple Mendelian genetics

- The relationship between genotype and phenotype is rarely as simple as in the pea plant characters Mendel studied
- Many heritable characters are not determined by only one gene with two alleles
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance

Extending Mendelian Genetics for a Single Gene

- Inheritance of characters by a single gene may deviate from simple Mendelian patterns in the following situations:
  - When alleles are not completely dominant or recessive
  - When a gene has more than two alleles
  - When a gene produces multiple phenotypes

Degrees of Dominance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In **incomplete dominance**, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
- In **codominance**, two dominant alleles affect the phenotype in separate, distinguishable ways

Fig. 14-10-1

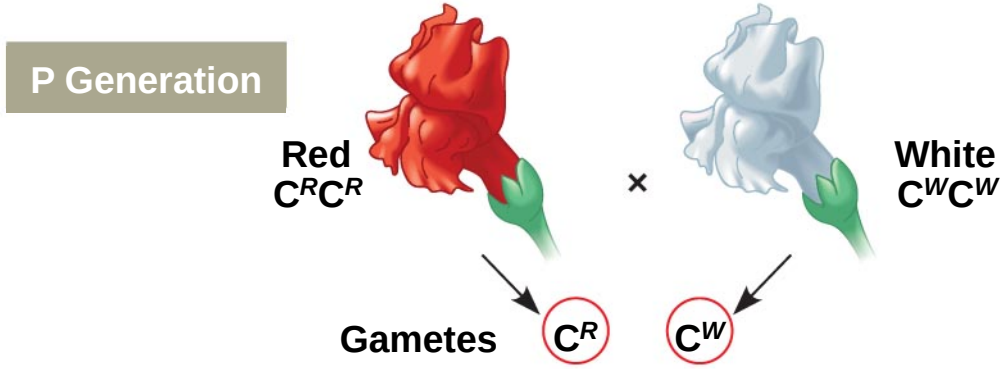


Fig. 14-10-2

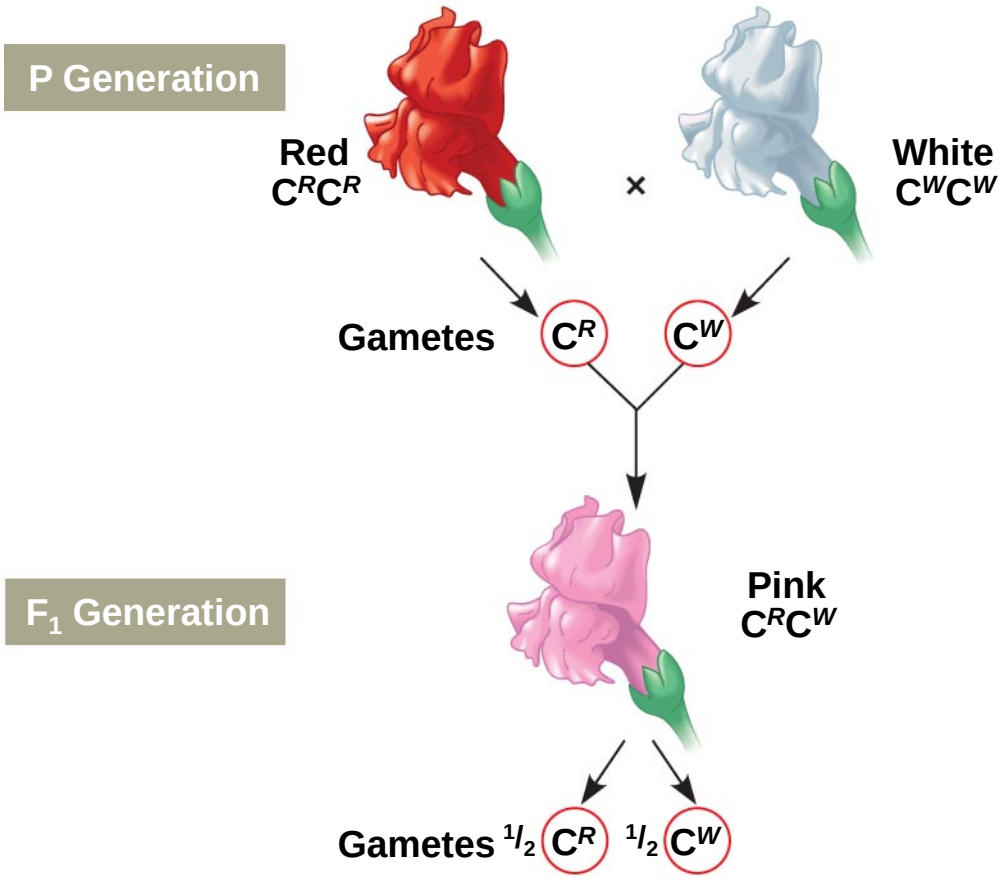
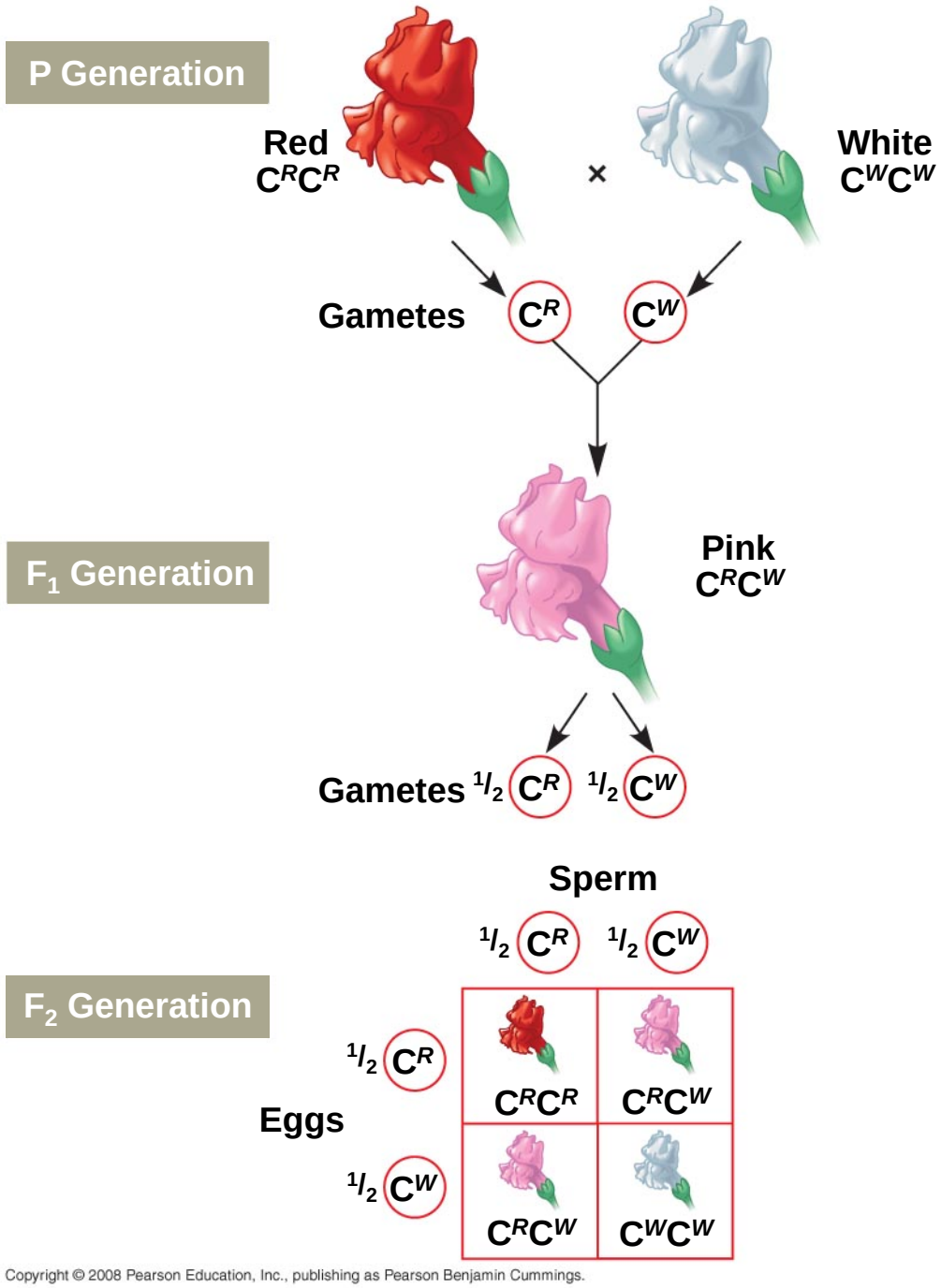


Fig. 14-10-3



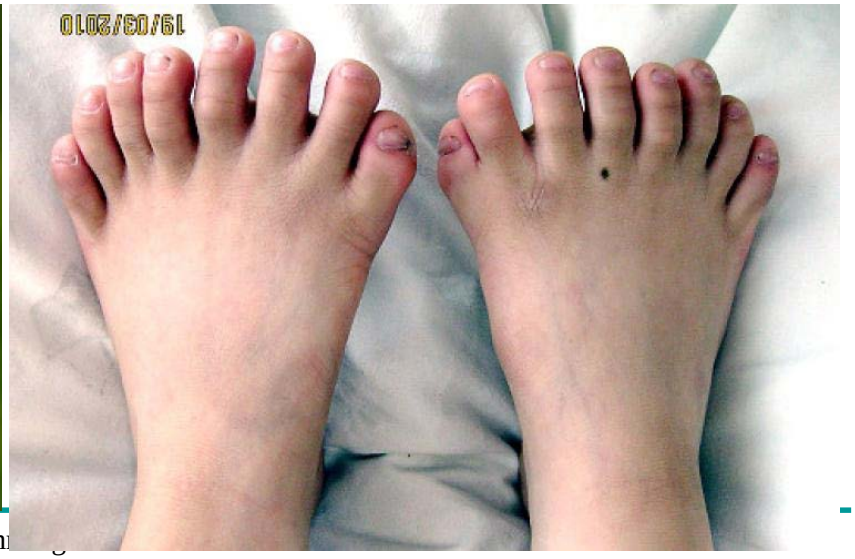
The Relation Between Dominance and Phenotype

- A dominant allele does not subdue a recessive allele; alleles don't interact
- Alleles are simply variations in a gene's nucleotide sequence
- For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype

- **Tay-Sachs disease** is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
  - At the *organismal* level, the allele is recessive
  - At the *biochemical* level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
  - At the *molecular* level, the alleles are codominant

Frequency of Dominant Alleles

- Dominant alleles are not necessarily more common in populations than recessive alleles
- For example, one baby out of 400 in the United States is born with extra fingers or toes



- The allele for this unusual trait is dominant to the allele for the more common trait of five digits per appendage
- In this example, the recessive allele is far more prevalent than the population's dominant allele

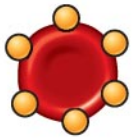
Multiple Alleles

- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells: I^A, I^B, and *i*.
- The enzyme encoded by the I^A allele adds the A carbohydrate, whereas the enzyme encoded by the I^B allele adds the B carbohydrate; the enzyme encoded by the *i* allele adds neither

Fig. 14-11

Allele	Carbohydrate
I^A	A
I^B	B
i	none

(a) The three alleles for the ABO blood groups and their associated carbohydrates

Genotype	Red blood cell appearance	Phenotype (blood group)
$I^A I^A$ or $I^A i$		A
$I^B I^B$ or $I^B i$		B
$I^A I^B$		AB
ii		O

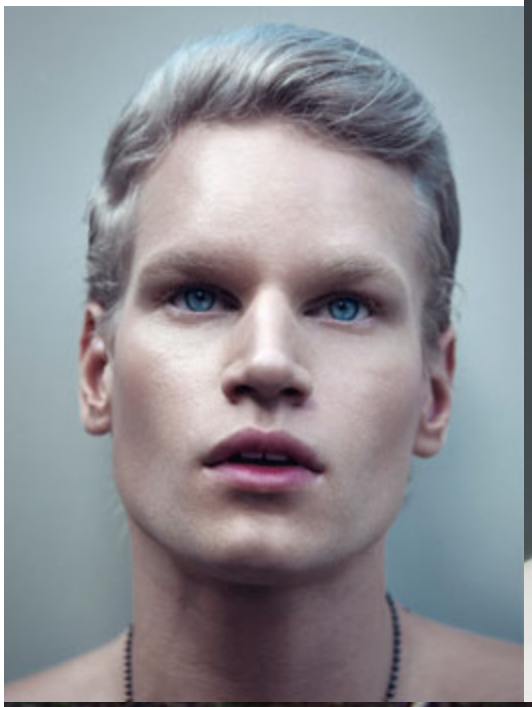
(b) Blood group genotypes and phenotypes

Pleiotropy

- Most genes have multiple phenotypic effects, a property called **pleiotropy**
- For example: Albinism
- One mutated gene for one enzyme causes albinism. Melanin production is little to none. There is no brown pigment in the eyes, hair, or skin.

Extending Mendelian Genetics for Two or More Genes

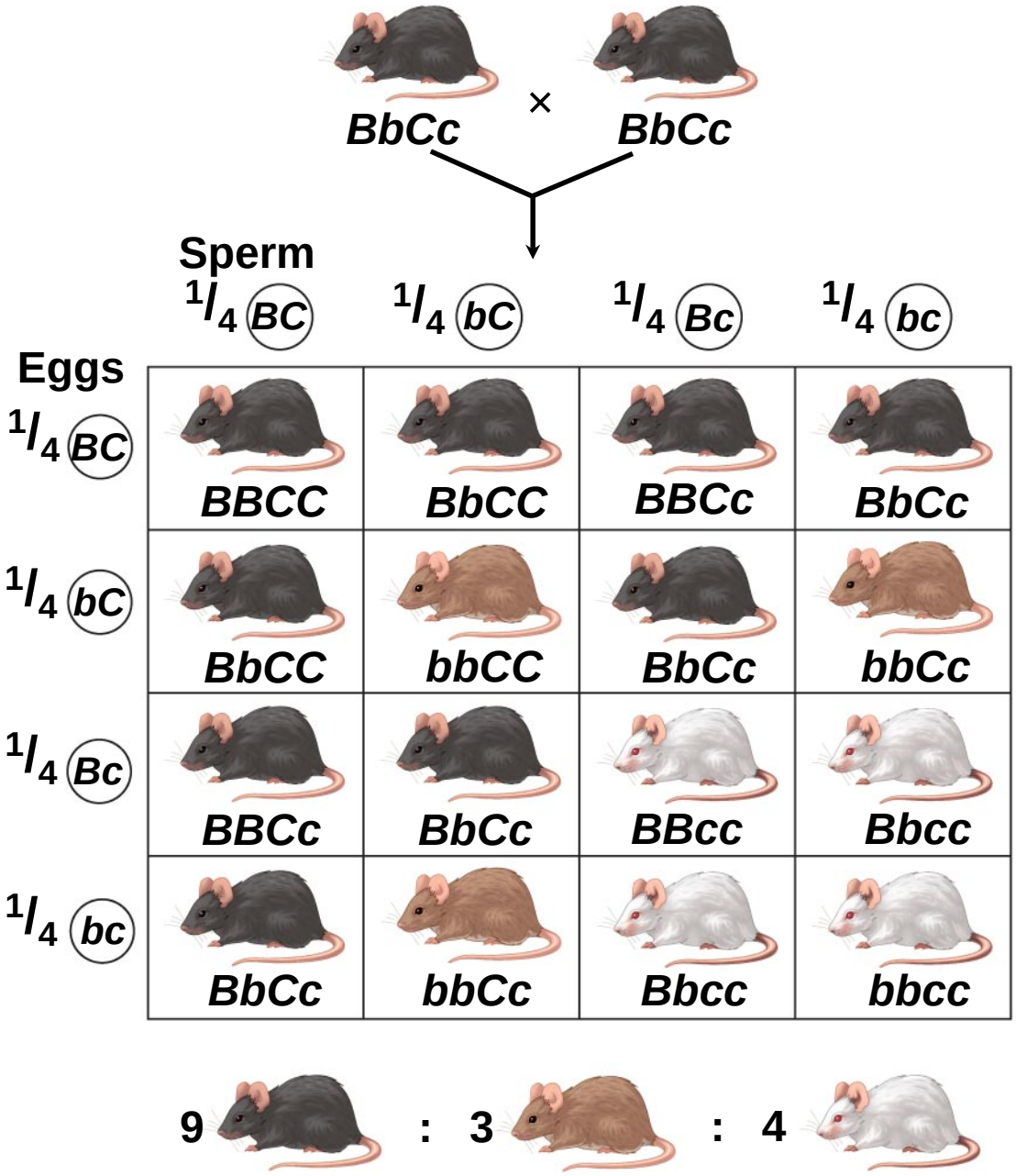
- Some traits may be determined by two or more genes



Epistasis

- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus (modifier genes)
- Examples: albinism, baldness, and in mice and many other mammals, coat color depends on two genes
- One gene determines the pigment color (with alleles *B* for black and *b* for brown)
- The other gene (with alleles *C* for color and *c* for no color) determines whether the pigment will be deposited in the hair

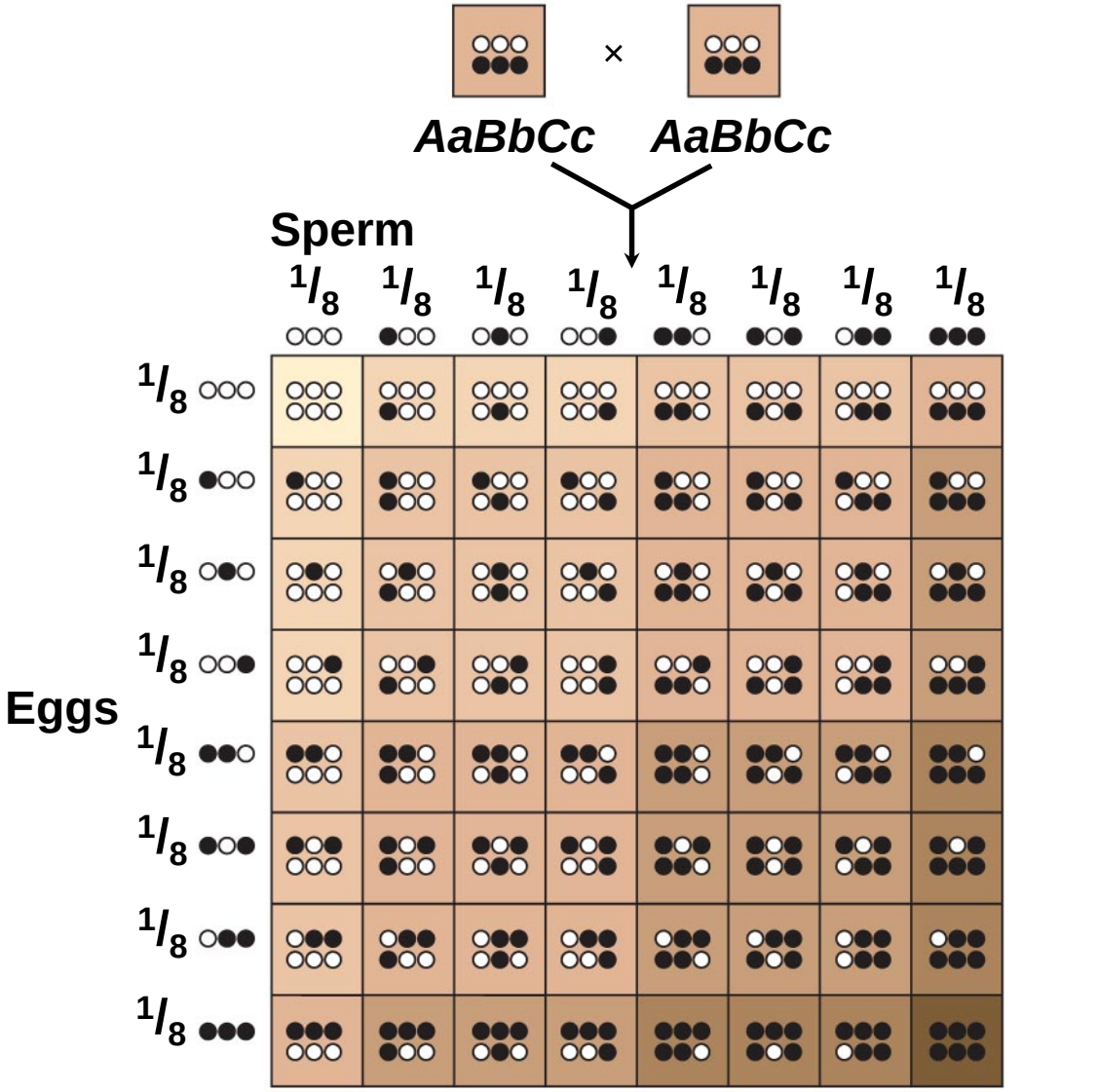
Fig. 14-12



Polygenic Inheritance

- **Quantitative characters** are those that vary in the population along a continuum
- Quantitative variation usually indicates **polygenic inheritance**, an additive effect of two or more genes on a single phenotype
- Skin color in humans is an example of polygenic inheritance

Fig. 14-13



Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype
- The **norm of reaction** is the phenotypic range of a genotype influenced by the environment
- For example, hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity

Fig. 14-14

- Norms of reaction are generally broadest for polygenic characters
- Such characters are called **multifactorial** because genetic and environmental factors collectively influence phenotype

Integrating a Mendelian View of Heredity and Variation

- An organism's phenotype includes its physical appearance, internal anatomy, physiology, and behavior
- An organism's phenotype reflects its overall genotype and unique environmental history

Concept 14.4: Many human traits follow Mendelian patterns of inheritance

- Humans are not good subjects for genetic research
  - Generation time is too long
  - Parents produce relatively few offspring
  - Breeding experiments are unacceptable
- However, basic Mendelian genetics endures as the foundation of human genetics

Pedigree Analysis

- A **pedigree** is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees
- Pedigrees can also be used to make predictions about future offspring

Fig. 14-15a

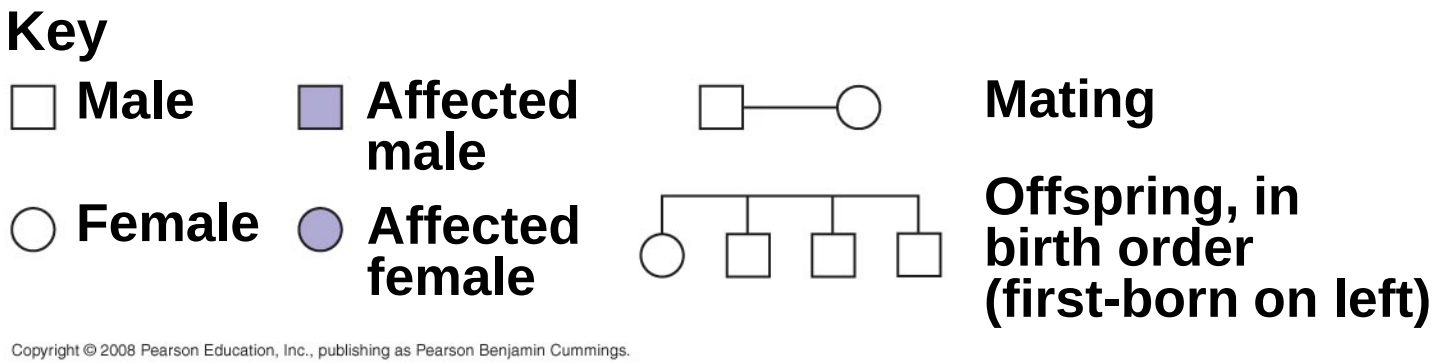
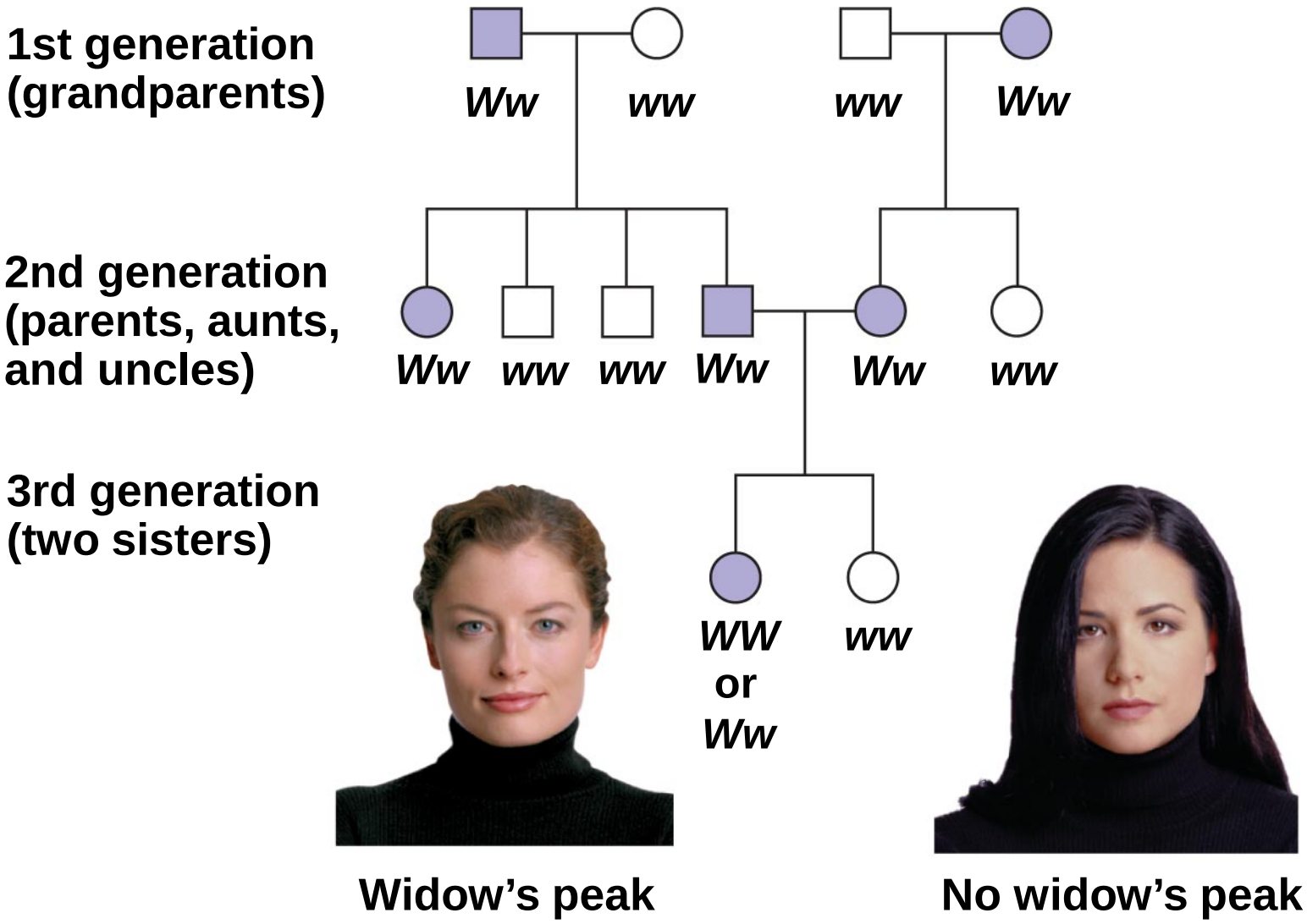
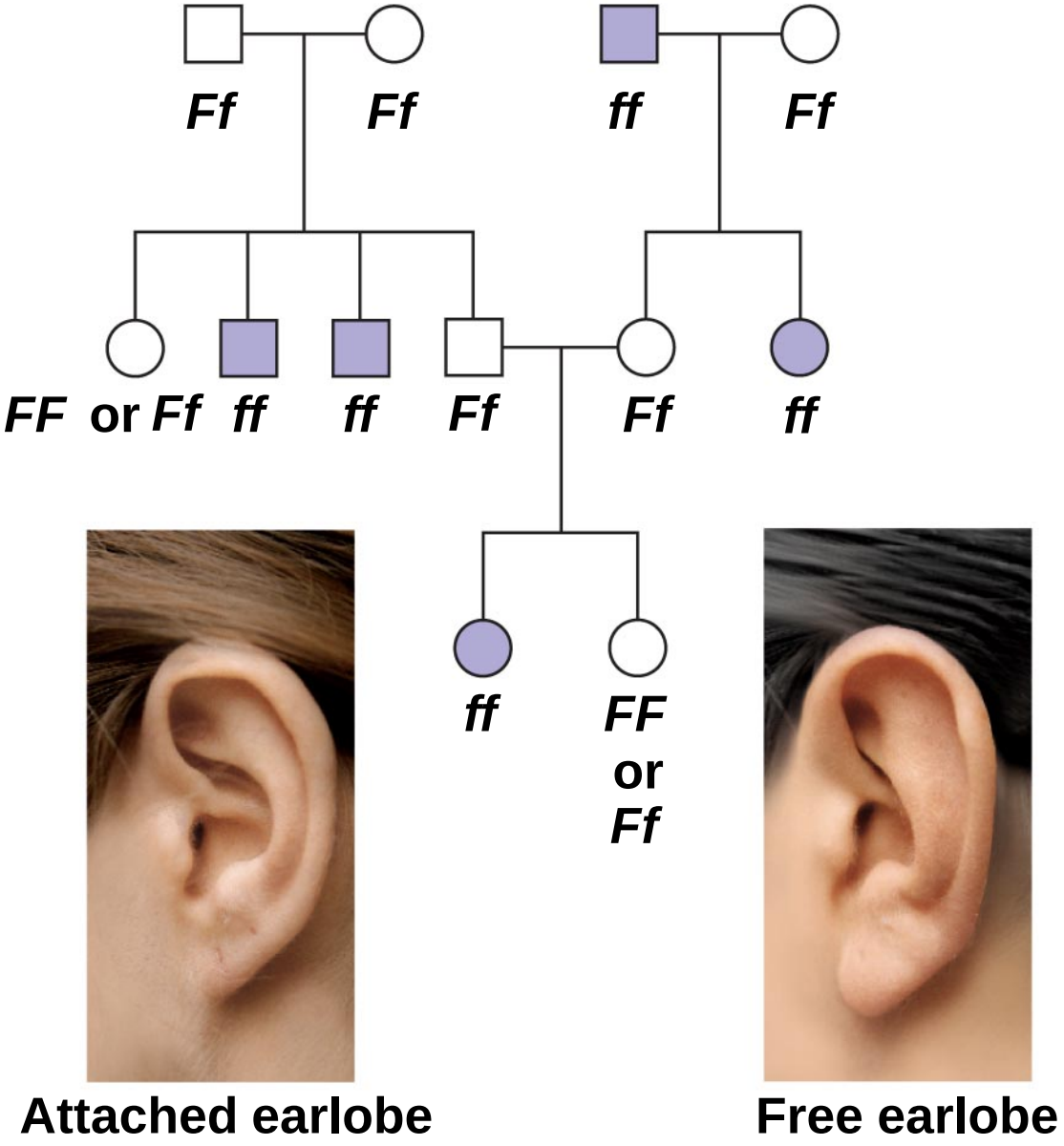


Fig. 14-15b



(a) Is a widow's peak a dominant or recessive trait?

Fig. 14-15c



(b) Is an attached earlobe a dominant or recessive trait?

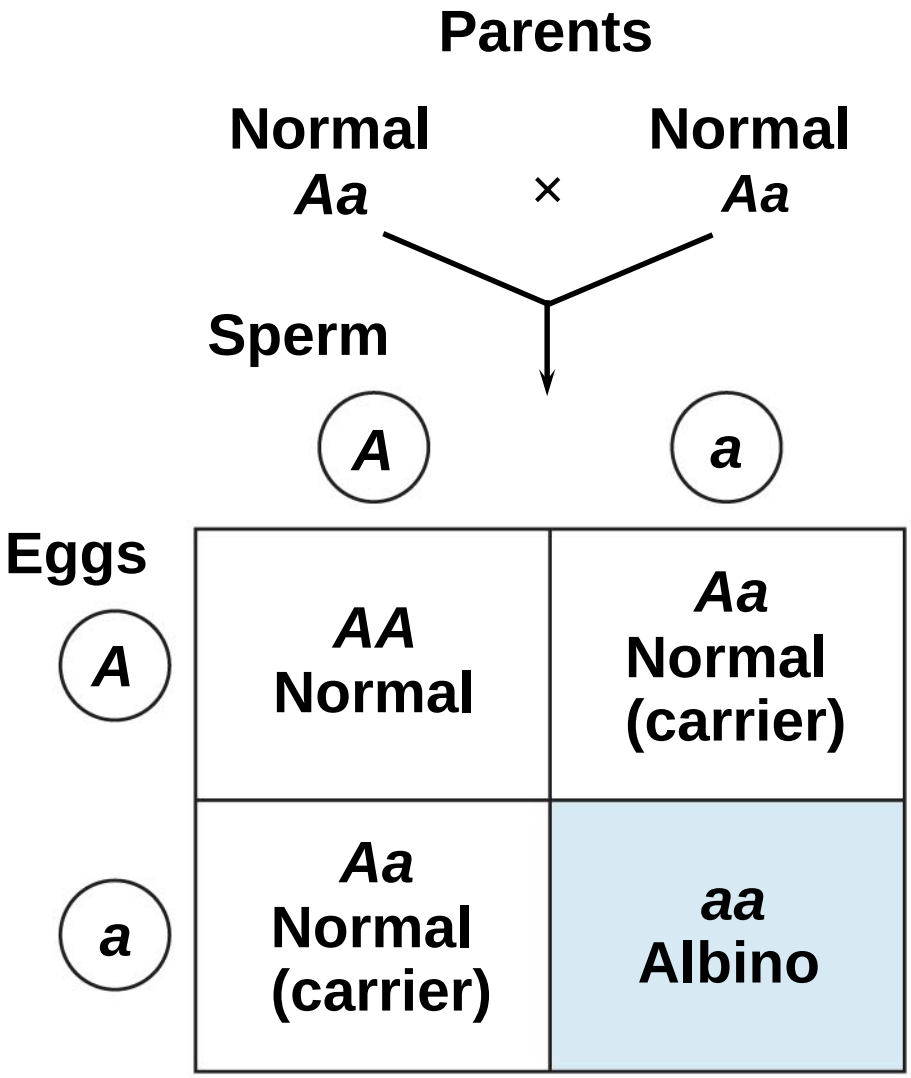
Recessively Inherited Disorders

- Many genetic disorders are inherited in a recessive manner

The Behavior of Recessive Alleles

- Recessively inherited disorders show up only in individuals homozygous for the allele
- **Carriers** are heterozygous individuals who carry the recessive allele but are phenotypically normal (i.e., pigmented)
- Albinism is a recessive condition characterized by a lack of pigmentation in skin, hair, and eyes.

Fig. 14-16

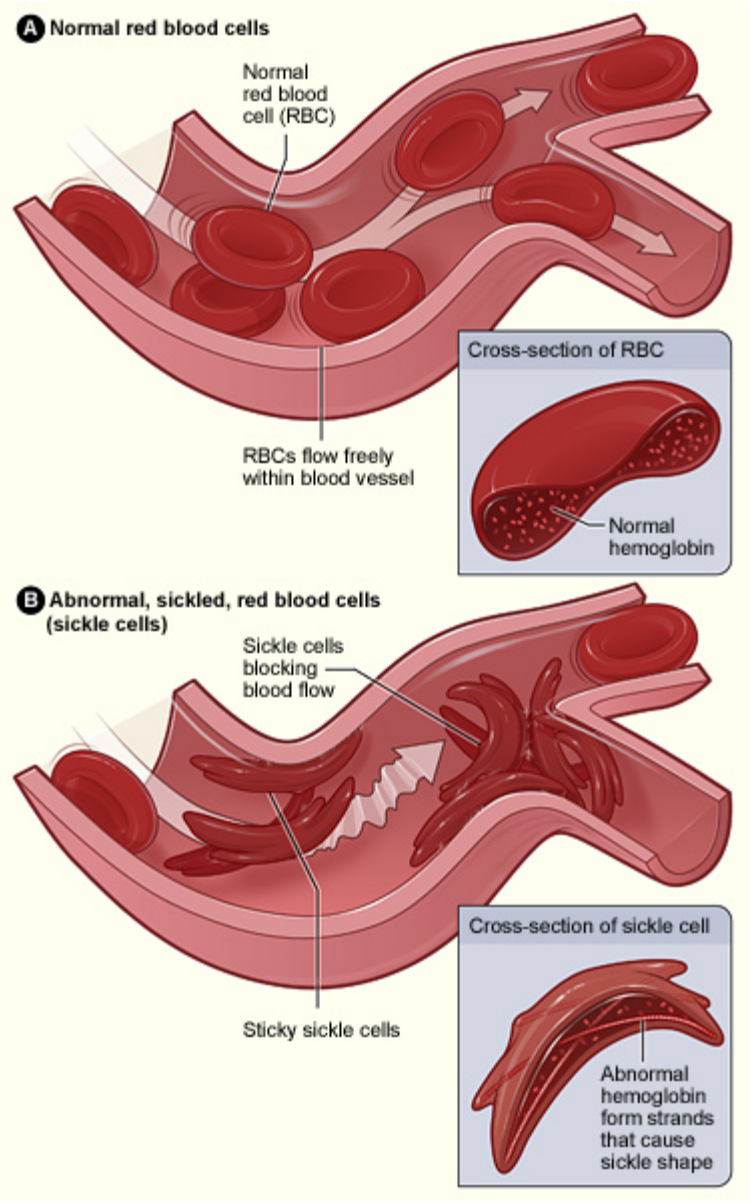


- If a recessive allele that causes a disease is rare, then the chance of two carriers meeting and mating is low
- Consanguineous matings (i.e., matings between close relatives) increase the chance of mating between two carriers of the same rare allele
- Most societies and cultures have laws or taboos against marriages between close relatives

Sickle-Cell Disease

- **Sickle-cell disease** affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- Symptoms include physical weakness, pain, organ damage, and even paralysis
- The sickle cell trait evolved for a purpose!!!!!!

- As of 2015 about 4.4 million people have sickle-cell disease while an additional 43 million have sickle-cell trait
- ~ 80% of sickle-cell disease cases are believed to occur in sub-Saharan Africa
- In 2015, it resulted in about 114,800 deaths
- So, why did this trait evolve?

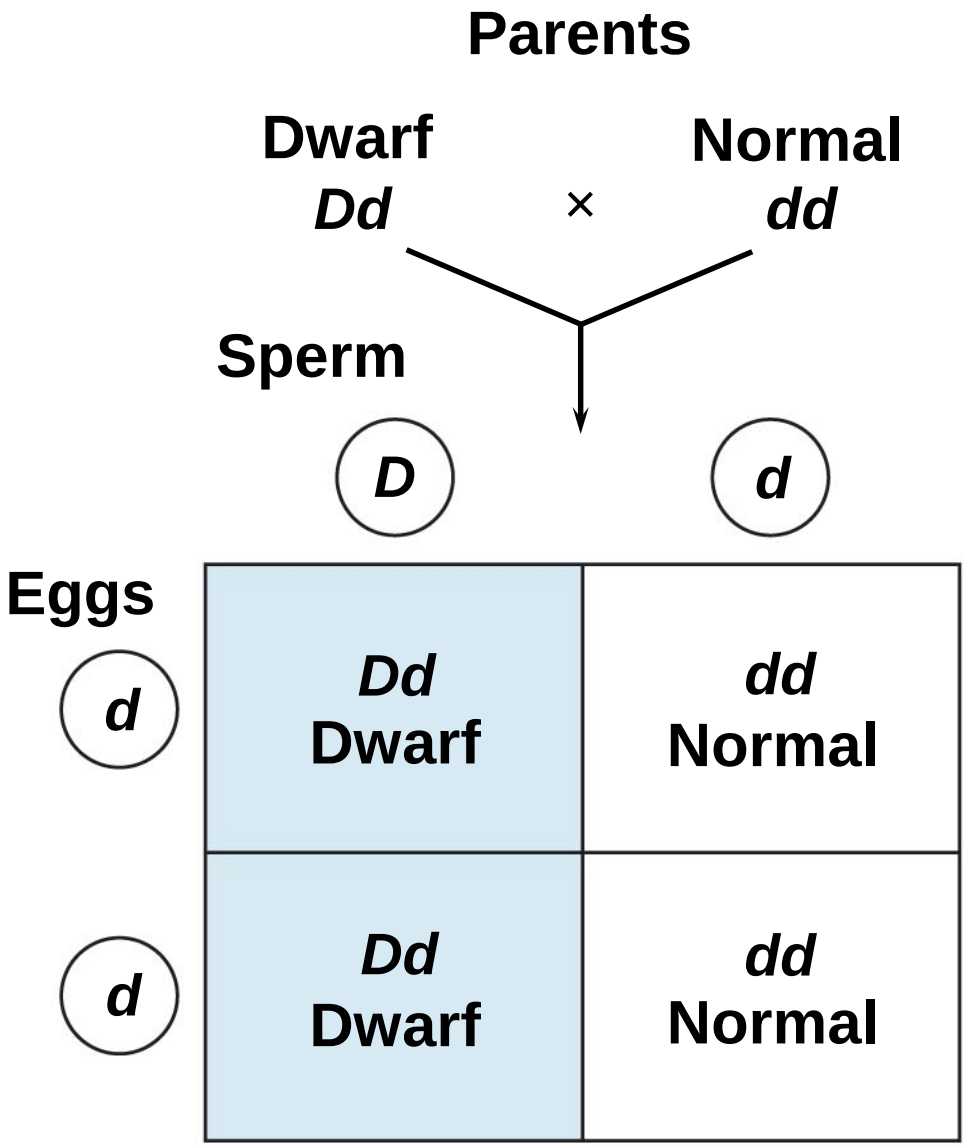


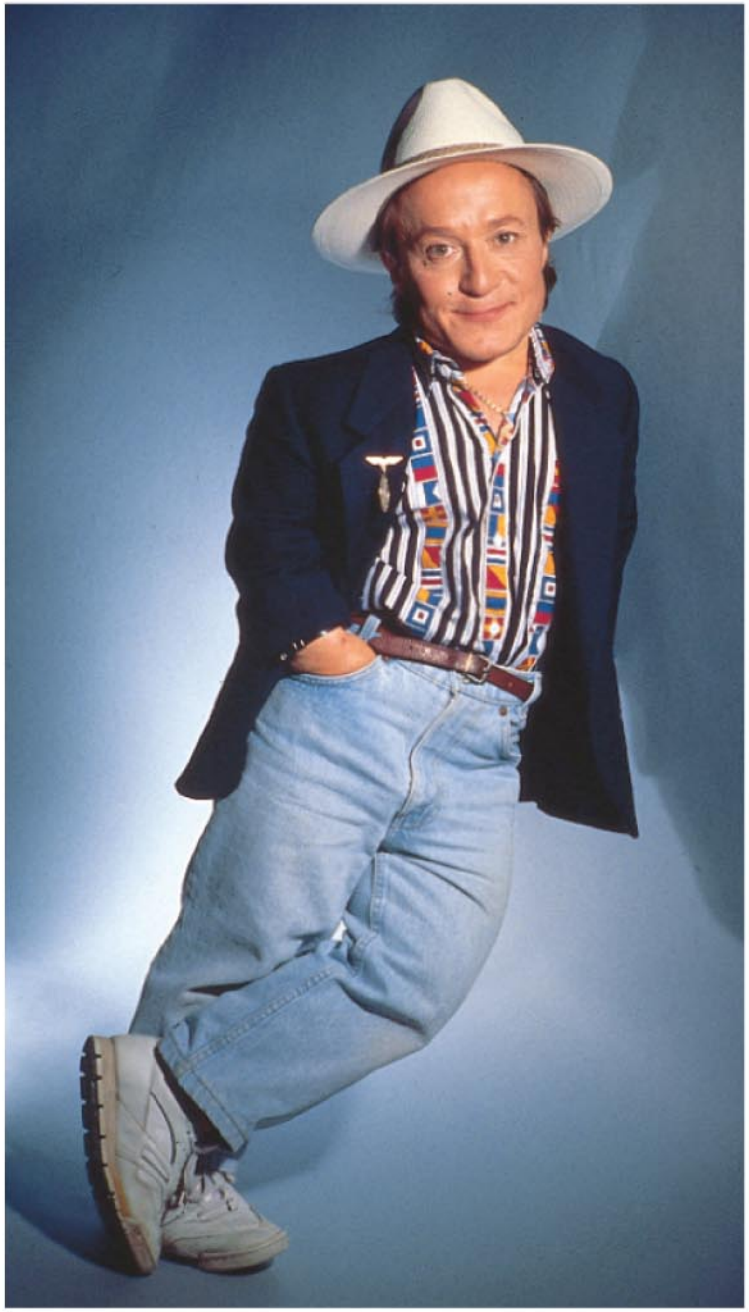
- Being heterozygous for sickle cell results in protection from malaria.
- Heterozygotes have greater survival in areas where malaria is endemic (always present).
- Unfortunately, being homozygous for sickle cell increases risk of death!
- Do a Punnett square and see that the majority of a population will be protected.

Dominantly Inherited Disorders

- Some human disorders are caused by dominant alleles
- Dominant alleles that cause a lethal disease are rare and arise by mutation
- *Achondroplasia* is a form of dwarfism caused by a rare dominant allele

Fig. 14-17





Do a Punnett square where both parents have achondroplasia. What are the outcomes?

Huntington's Disease

- **Huntington's disease** is a degenerative disease of the nervous system
- The disease has no obvious phenotypic effects until the individual is about 35 to 40 years of age

Multifactorial Disorders

- Many diseases, such as heart disease and cancer, have both genetic and environmental components
- Little is understood about the genetic contribution to most multifactorial diseases

What to do?

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease
- Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders
- For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately

Newborn Screening

- Some genetic disorders can be detected in utero or at birth by simple tests that are now routinely performed in most hospitals in the United States

You should now be able to:

1. Define the following terms: true breeding, hybridization, monohybrid cross, P generation, F_1 generation, F_2 generation
2. Distinguish between the following pairs of terms: dominant and recessive; heterozygous and homozygous; genotype and phenotype
3. Use a Punnett square to predict the results of a cross and to state the phenotypic and genotypic ratios of the F_2 generation

4. Explain how phenotypic expression in the heterozygote differs with complete dominance, incomplete dominance, and codominance

5. Define and give examples of pleiotropy and epistasis

6. Explain why lethal dominant genes are much rarer than lethal recessive genes

7. Explain how carrier recognition, fetal testing, and newborn screening can be used in genetic screening and counseling