Chapter 14

Mendel and the Gene Idea

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

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Learning English

英英字典

專有名詞

www.dictionary.com

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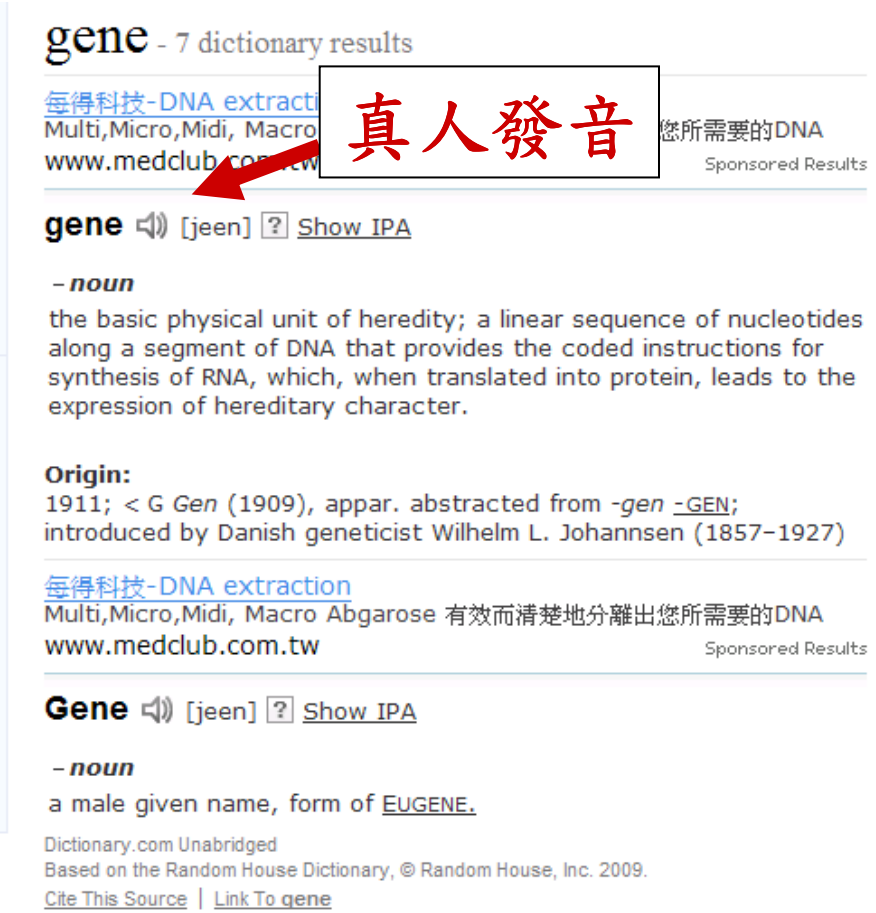
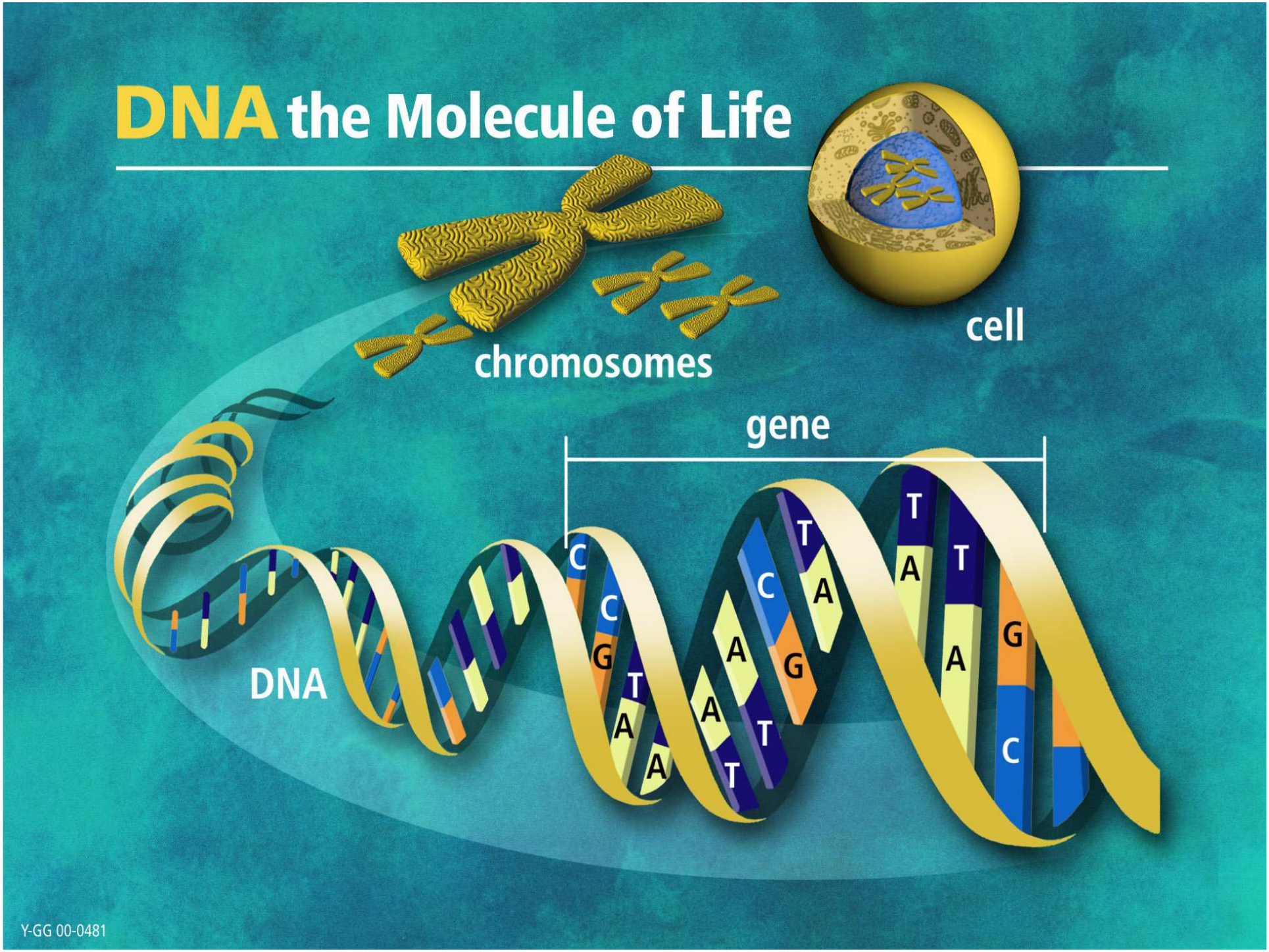


Campbell Biology Online Resource

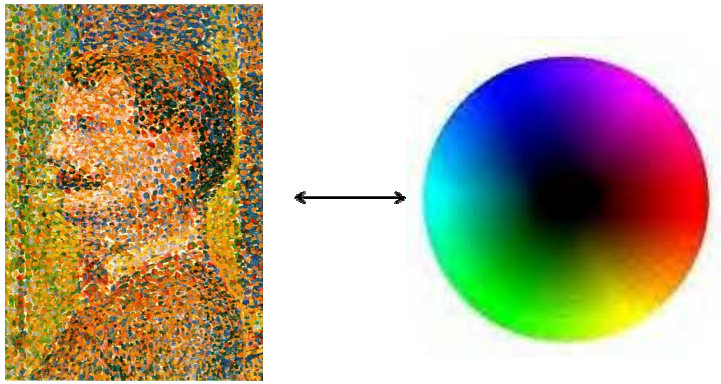
http://www.coursecompass.com/

MP3 tutor, Online lab, Word roots…


DNA the Molecule of Life
chromosomes
cell
gene
DNA
C
C
G
T
A
A
A
T
A
G
C
T
T
A
T
A
G
C
Y-GG 00-0481

Overview: Drawing from the Deck of Genes

- What genetic principles account for **the passing of traits** from parents to offspring?
- During the 1800s, the "blending" hypothesis is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)



The Gene idea!

- The “particulate” hypothesis (散粒; 不是 particular !) is the idea that parents pass on discrete (離散的;不連接的) heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas.

Fig. 14-1

Gregor Johann Mendel

[**greg**-er **yoh**-hann **men**-dl]

Mendelian [men-**dee**-lee-*uh*n]

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

If you were Mendel,

How will you design the experiment?

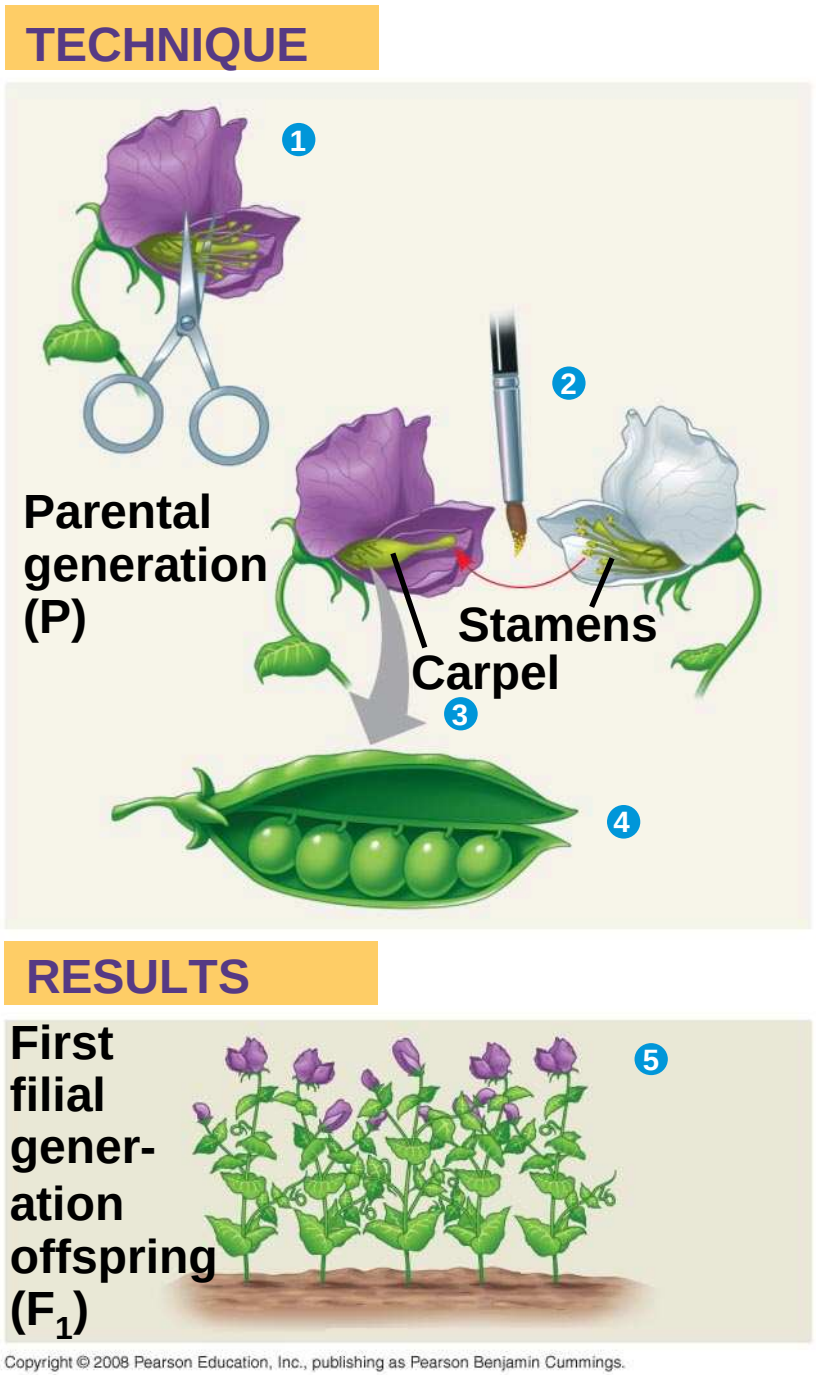
Mendel's Experimental, Quantitative Approach

- Advantages of pea plants for genetic study:
  - There are many varieties with distinct heritable features, or **characters** (such as flower color); character variants (such as purple or white flowers) are called **traits**
  - Mating of plants can be controlled
  - Each pea plant has sperm-producing organs (stamens) and egg-producing organs (carpels)
  - Cross-pollination (fertilization between different plants) can be achieved by dusting one plant with pollen from another

Fig. 14-2

P: Parental

F: Filial



Selecting the experimental variable/parameter

- Mendel chose to track only those characters that varied in an either-or manner (either-or; 非白即紫)
- He also used varieties that were **true-breeding** (plants that produce offspring of the same variety when they self-pollinate)

Hybridization 雜交

- 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

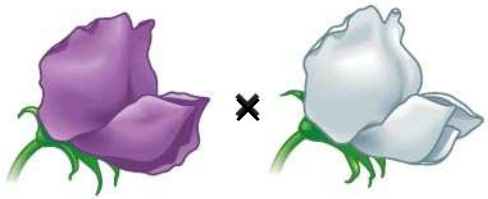
The Law of Segregation

- 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)

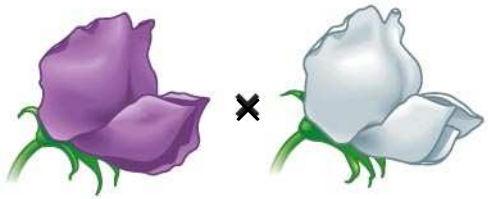


Purple flowers × **White flowers**

Fig. 14-3-2

EXPERIMENT

P Generation (true-breeding parents)

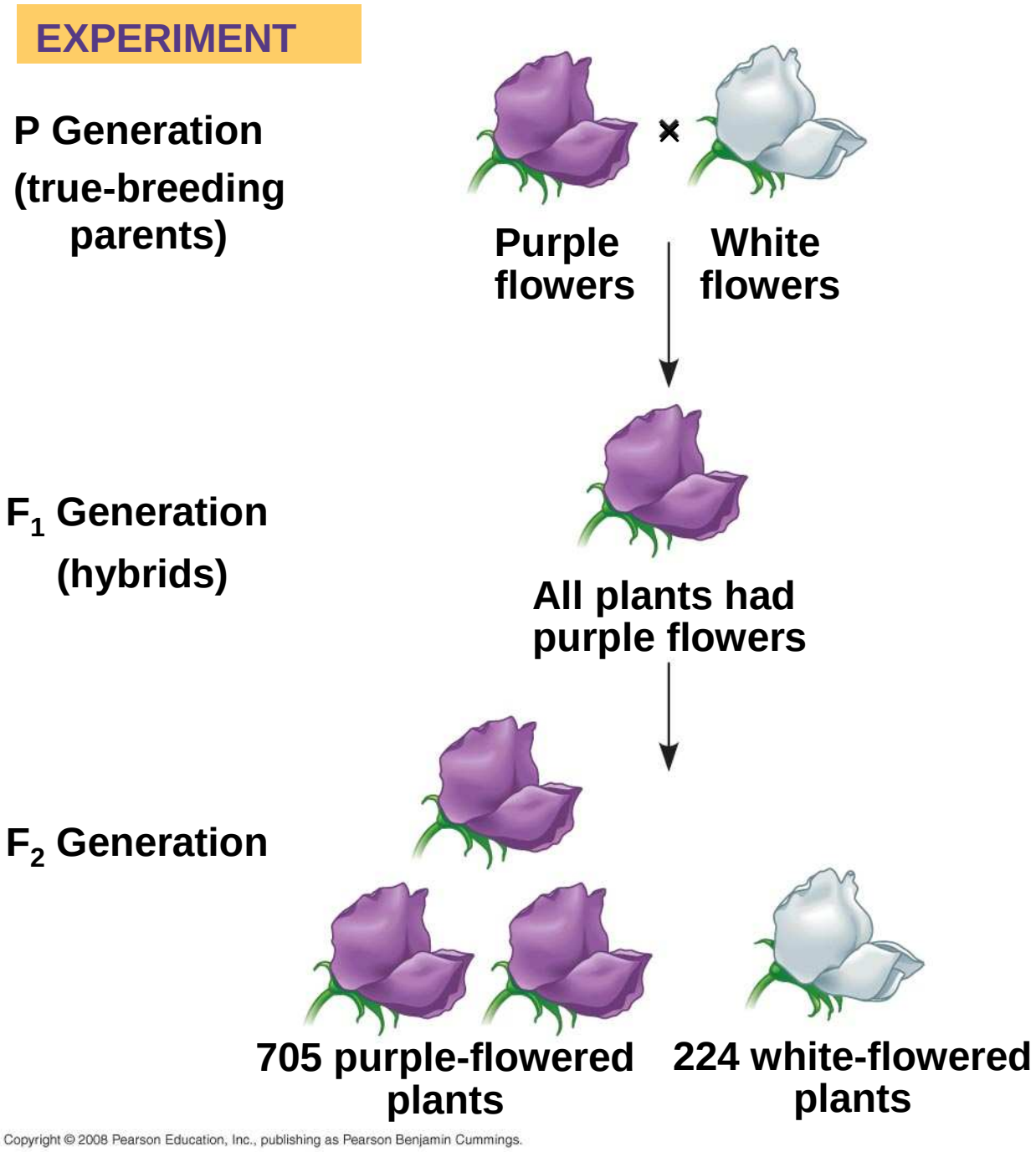


Purple flowers × **White flowers**

F_1 Generation (hybrids)

All plants had purple flowers

Fig. 14-3-3



Data Analysis and Interpretation

- 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
- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits
- What Mendel called a "heritable factor" is what we now call a gene

Table 14-1

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	x	Recessive Trait	F_2 Generation Dominant:Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

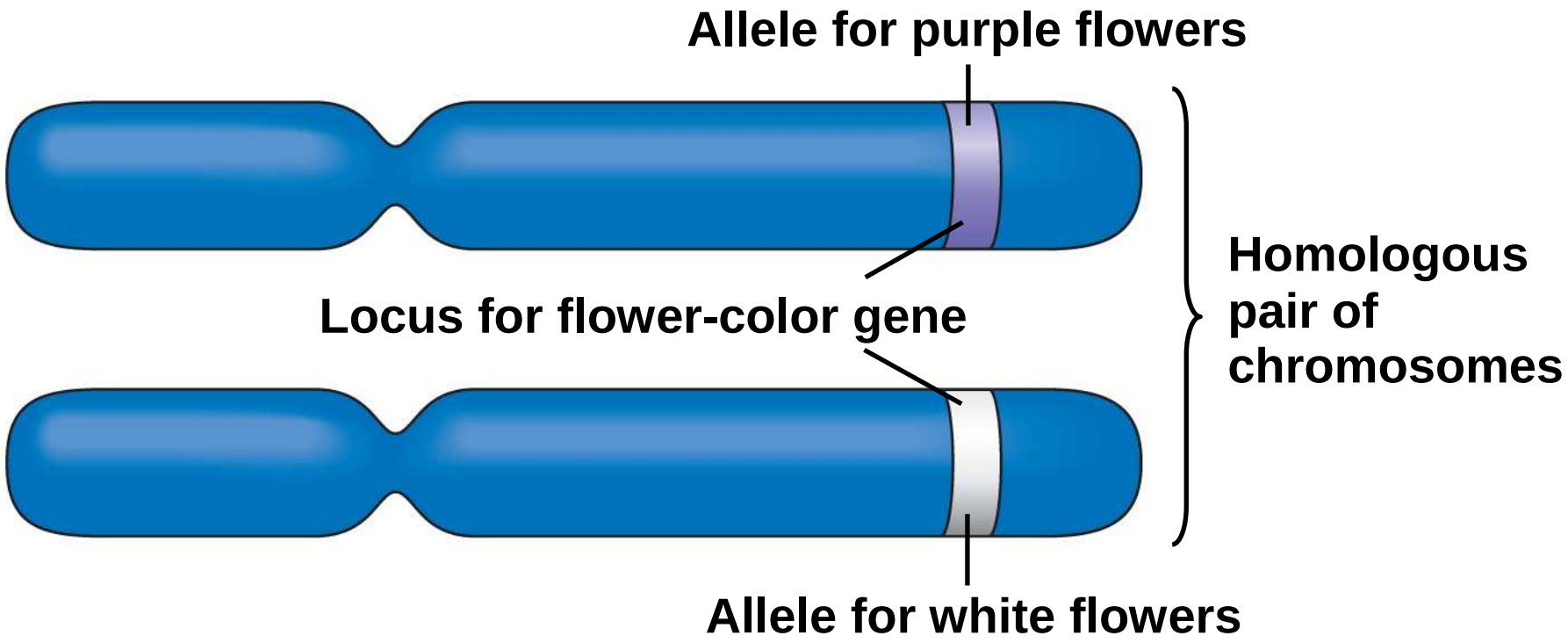
Mendel's Model – hypothesis to explain the data

- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F_2 offspring
- Four related concepts make up this model
- These concepts can be related to what we now know about genes and chromosomes

(1/4) Alleles: alternative versions of genes

- 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



(2/4) Each parent offers one allele

- 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

(3/4) Dominant vs. Recessive allele

- 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

(4/4) One heritable character in one gamete

- 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 the body) of an organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes (sperm or egg) in meiosis

Punnett square

- 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

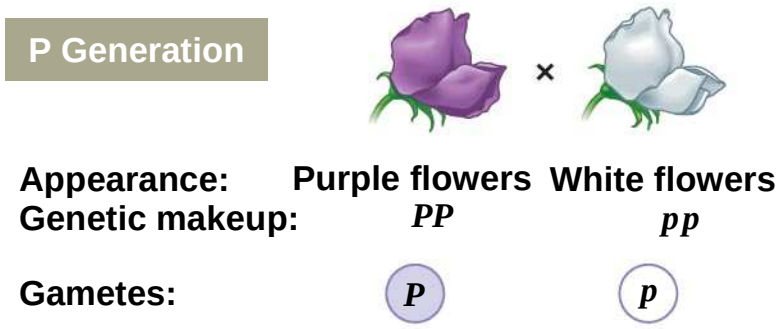


Fig. 14-5-2

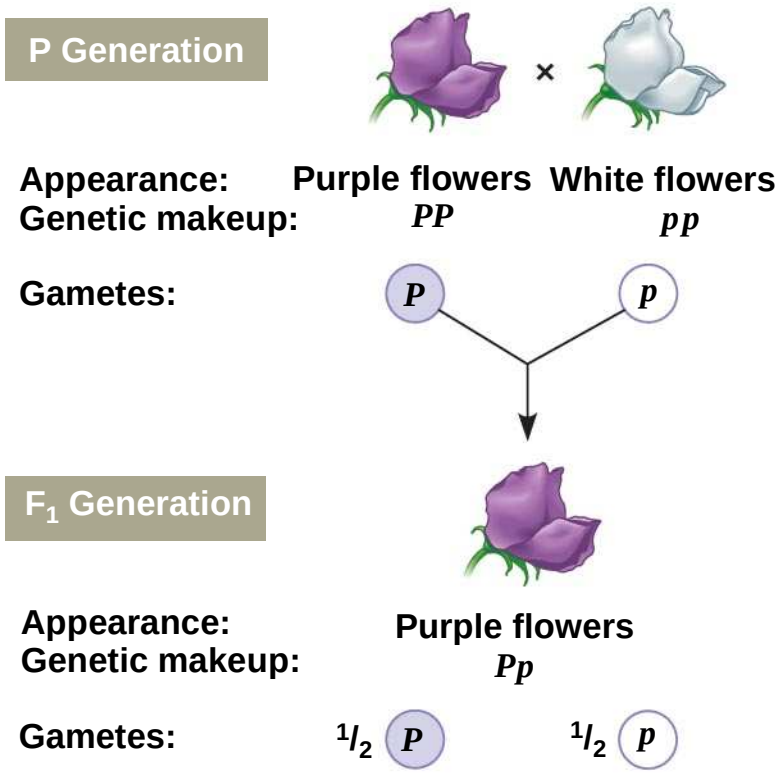
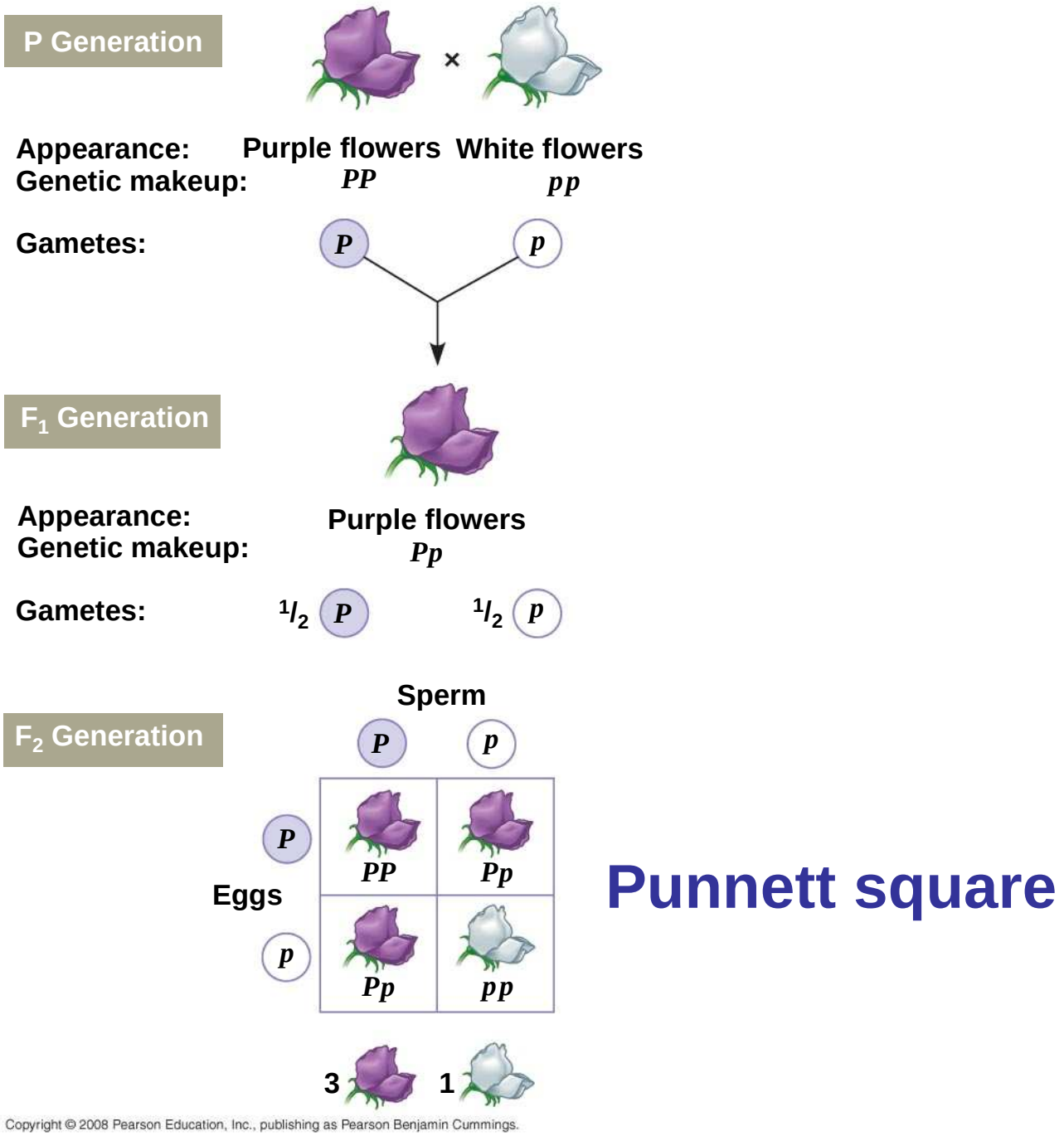


Fig. 14-5-3



Punnett square

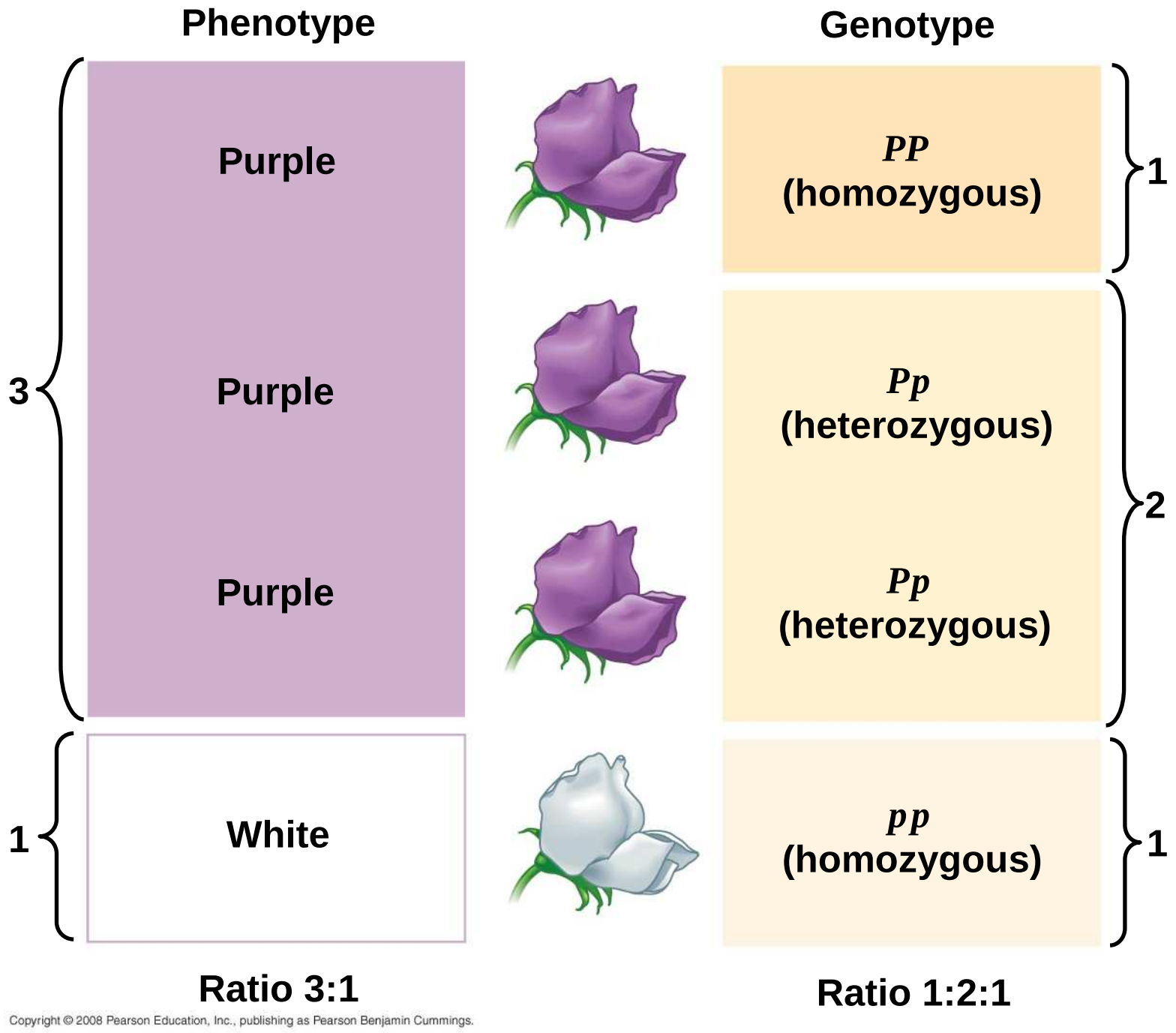
Useful Genetic Vocabulary

- 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

Phenotype vs. Genotype

- 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 (試交)

- 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. 14-7a

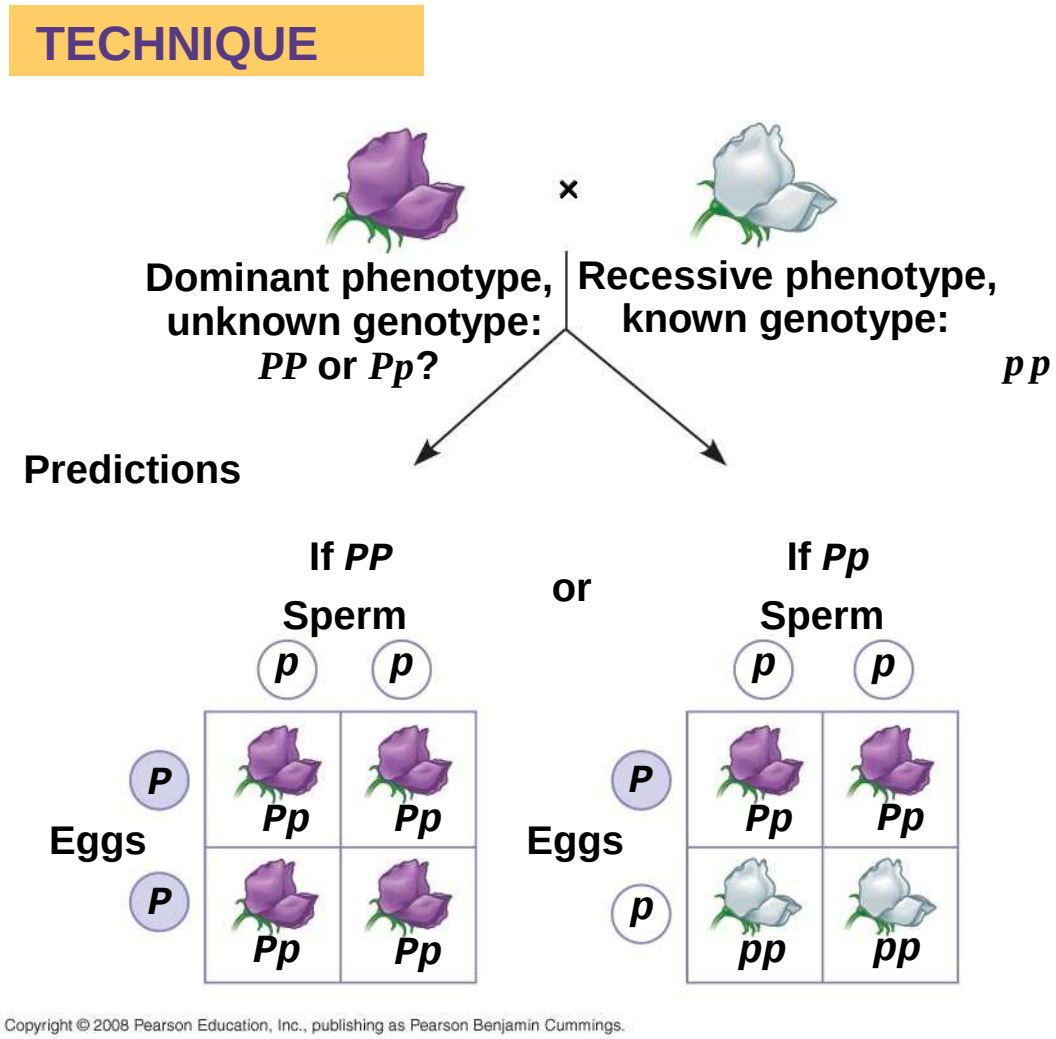


Fig. 14-7b

RESULTS

All offspring purple

or

$^1/_2$ offspring purple and $^1/_2$ offspring white

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*

Changing experimental condition

- 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. 14-8a

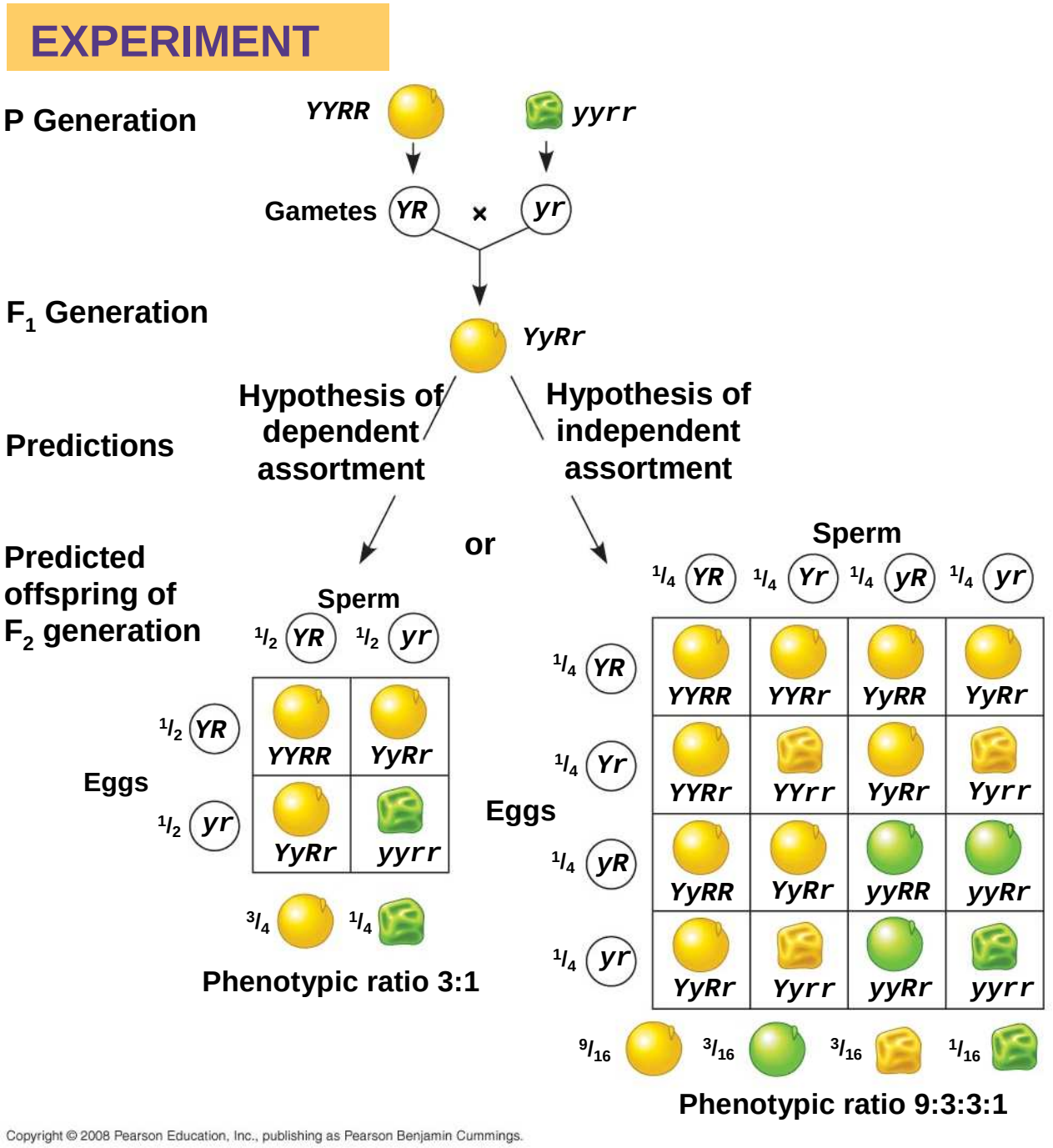


Fig. 14-8b

RESULTS

315  108  101  32 

Phenotypic ratio approximately 9:3:3:1

Law of independent assortment

- 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

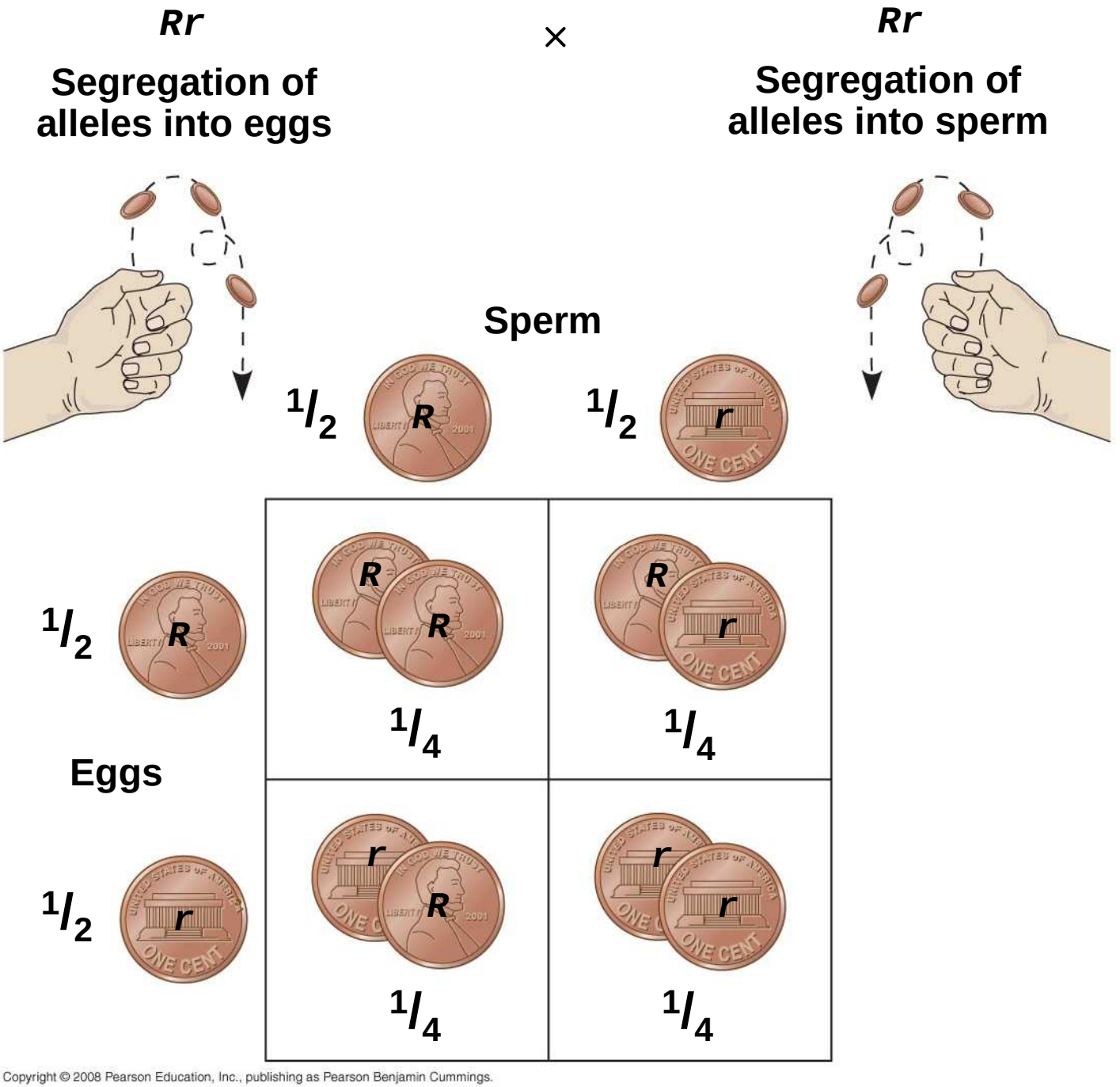
Concept 14.2: The laws of probability govern Mendelian inheritance

- 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

The Multiplication and Addition Rules Applied to Monohybrid Crosses

- The multiplication rule states that the probability that two or more independent events will occur together is the product of their individual probabilities
- 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

Fig. 14-9



Rule of addition

- The rule of addition states that the probability that any one of two or more exclusive events will occur is calculated by adding together their individual probabilities
- The rule of addition can be used to figure out the probability that an F_2 plant from a monohybrid cross will be heterozygous rather than homozygous

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

Fig. 14-UN1

ppyyRr	¼ (probability of *pp*) × ½ (*yy*) × ½ (*Rr*)	= 1/16
ppYyrr	¼ × ½ × ½	= 1/16
Ppyyrr	½ × ½ × ½	= 2/16
PPyyrr	¼ × ½ × ½	= 1/16
ppyyrr	¼ × ½ × ½	= 1/16
Chance of *at least two* recessive traits		**= 6/16 or 3/8**

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

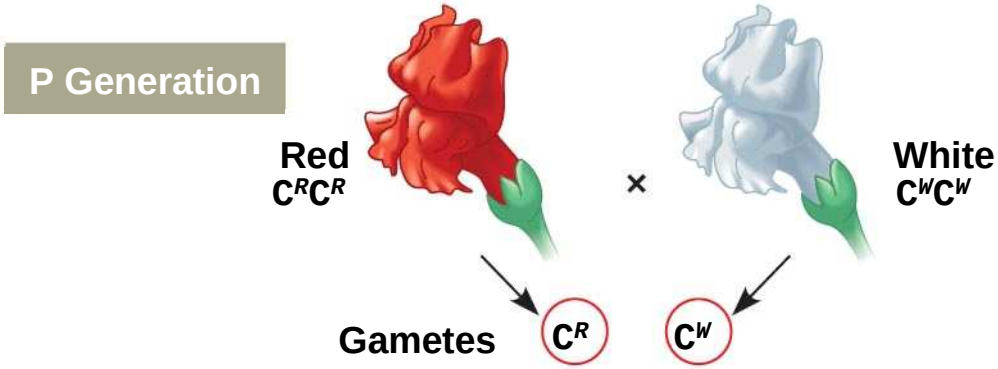


Fig. 14-10-2

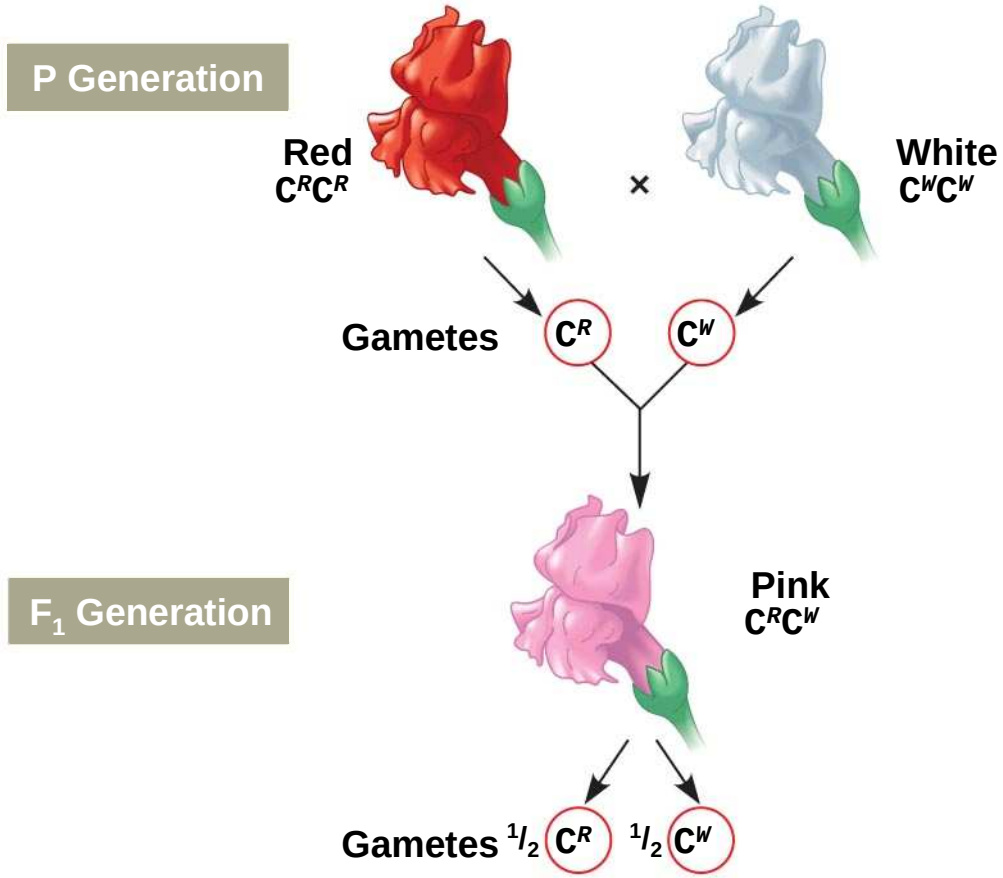
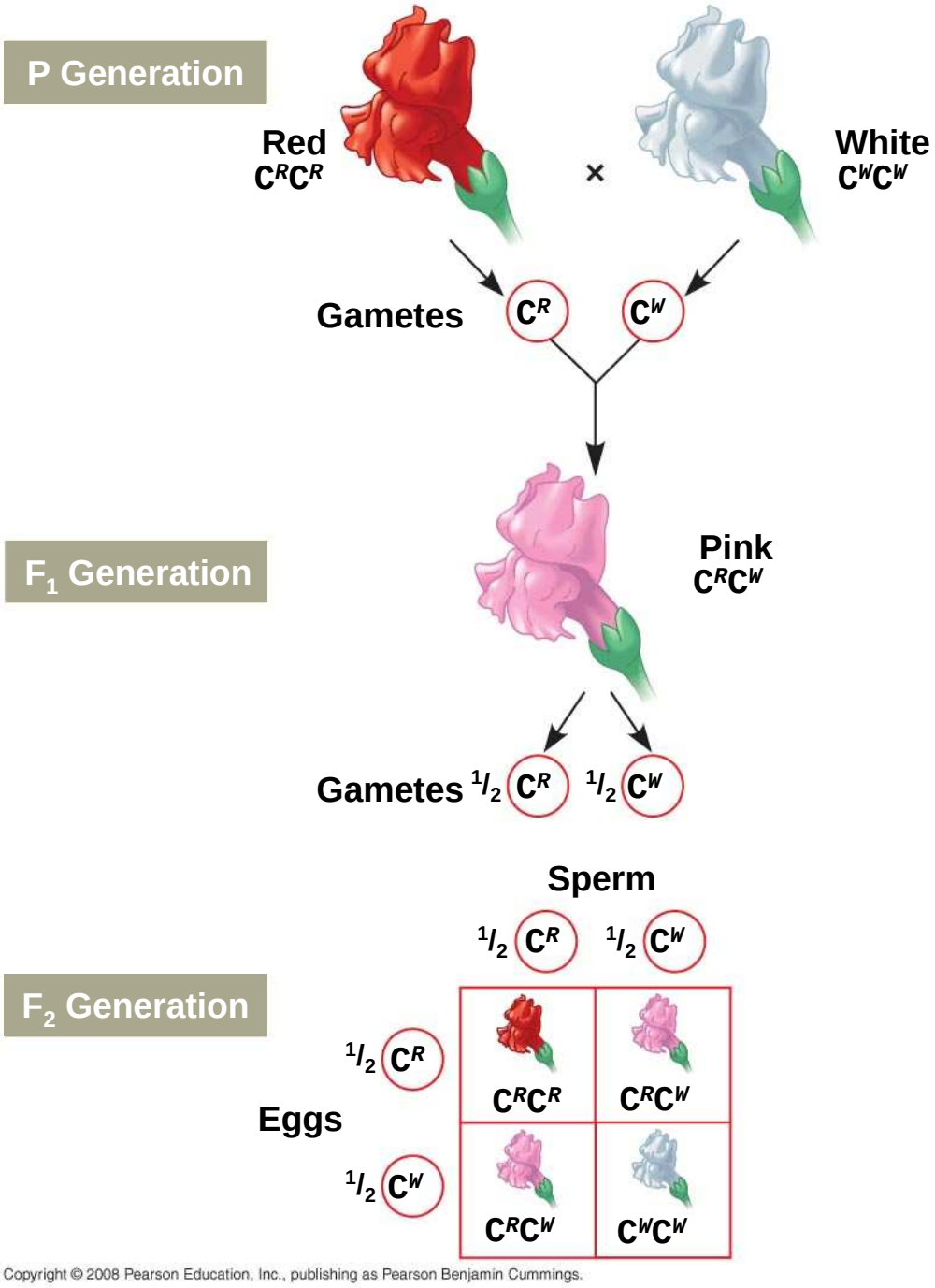


Fig. 14-10-3



Alleles are simply variations in a gene's sequence

The Relation Between Dominance and Phenotype

- A dominant allele does not subdue (壓制) 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 – varied degree of dominance at different levels

- **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

Do you have a extra finger or toe?

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

Prevalence (普及率；流行度) of a dominant allele

- 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

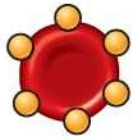
Multiple Alleles

- 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^AI^A or $I^A\,i$		A
I^BI^B or $I^B\,i$		B
I^AI^B		AB
ii		O

(b) Blood group genotypes and phenotypes

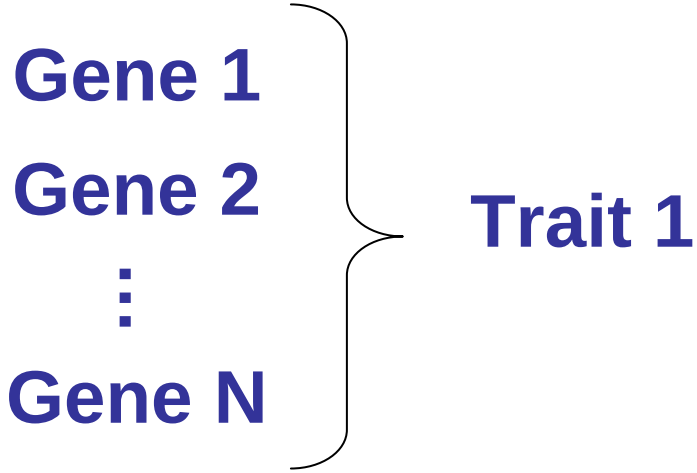
Pleiotropy (多重作用性)

- Most genes have multiple phenotypic effects, a property called **pleiotropy**
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as
  - cystic fibrosis（囊腫纖維症）
  - sickle-cell disease（鐮刀型細胞貧血）

名詞有超連結

Extending Mendelian Genetics for Two or More Genes

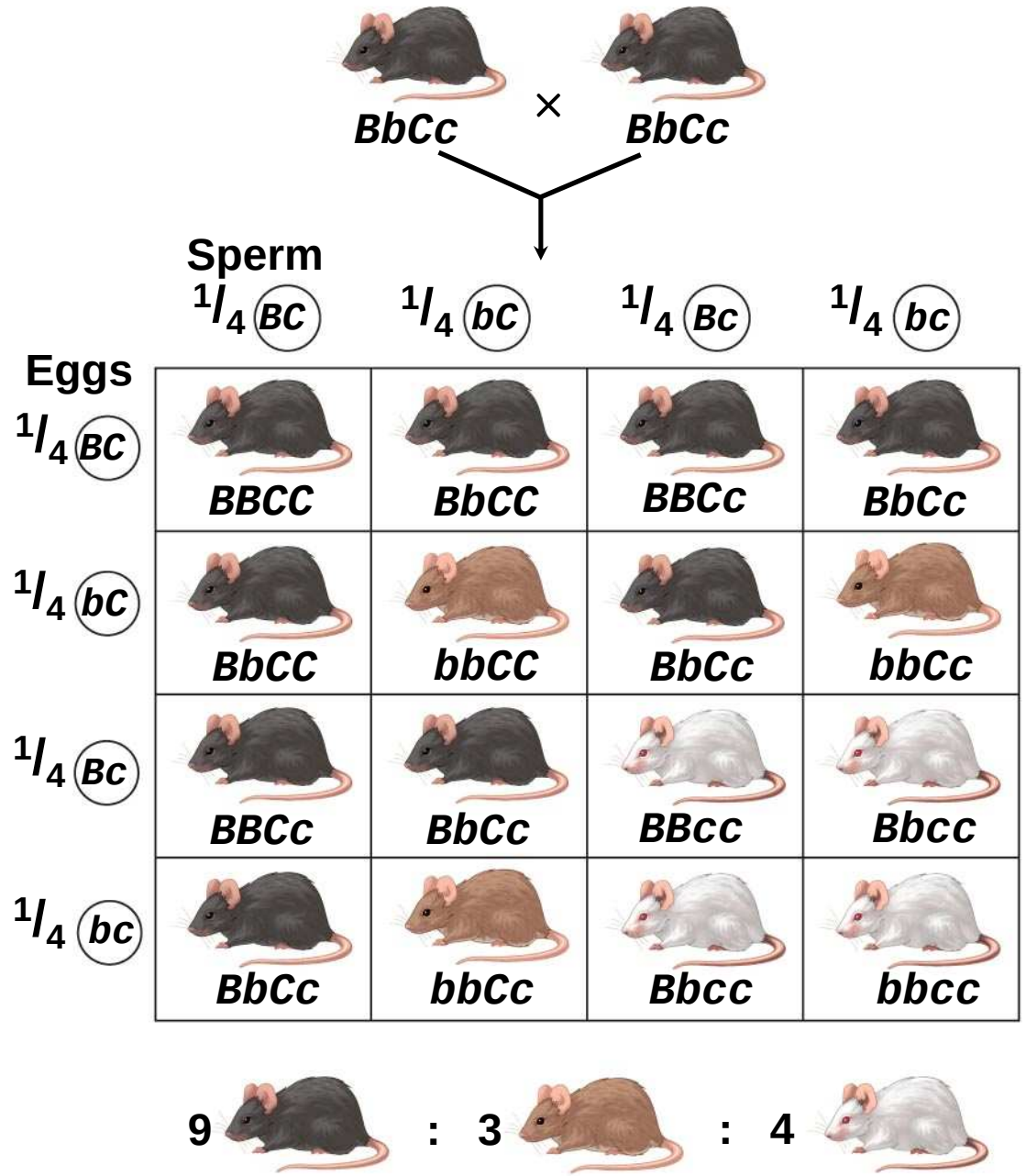
- Some traits may be determined by two or more genes



Epistasis (離位、越位調控)

- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, 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

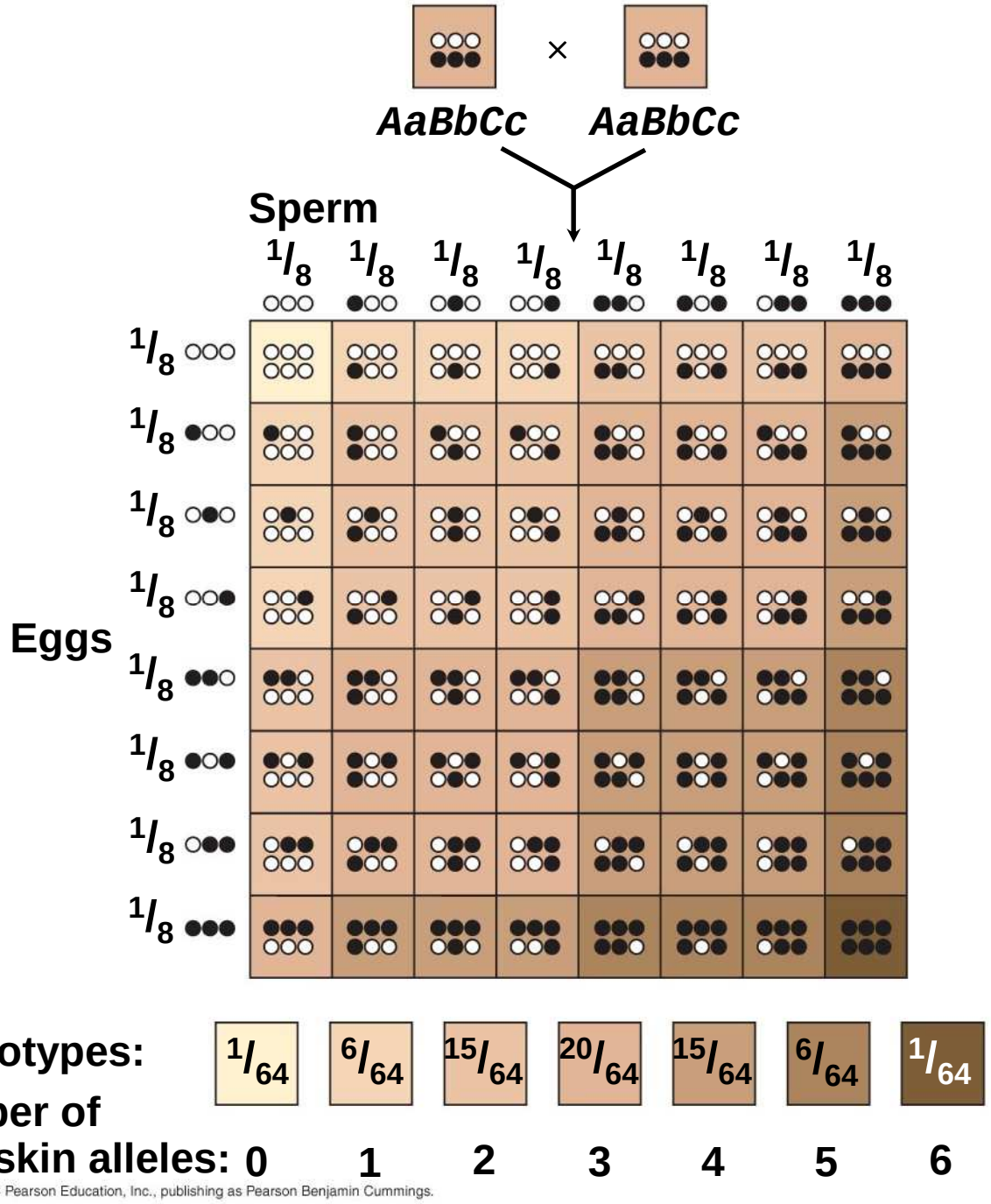
Fig. 14-12



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

繡球花 -- 花色可隨土壤的pH值而改變

若在酸性土壤種植(pH值比7小)，花色是藍色；若在中性土壤種植(pH值大約等於7)，花色是乳白色；若在鹼性土壤種植(pH值比7大)，花色是紅或紫 。

- 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 (族譜)

- 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

Fig. 14-15a

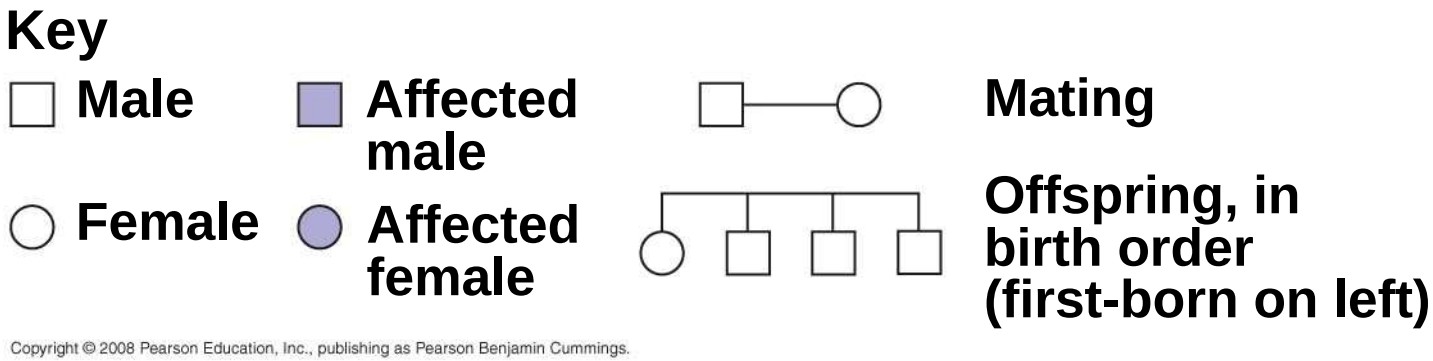
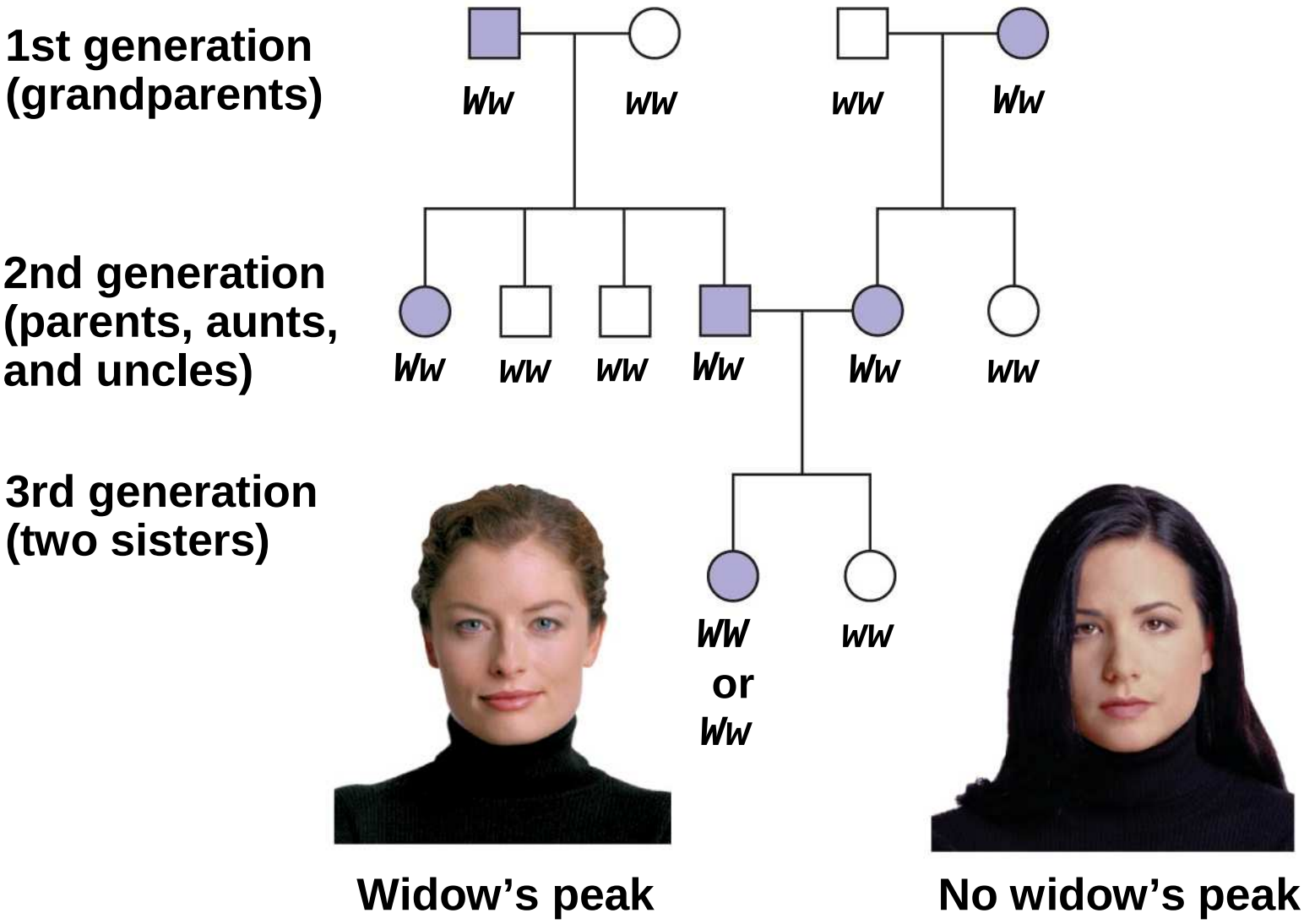
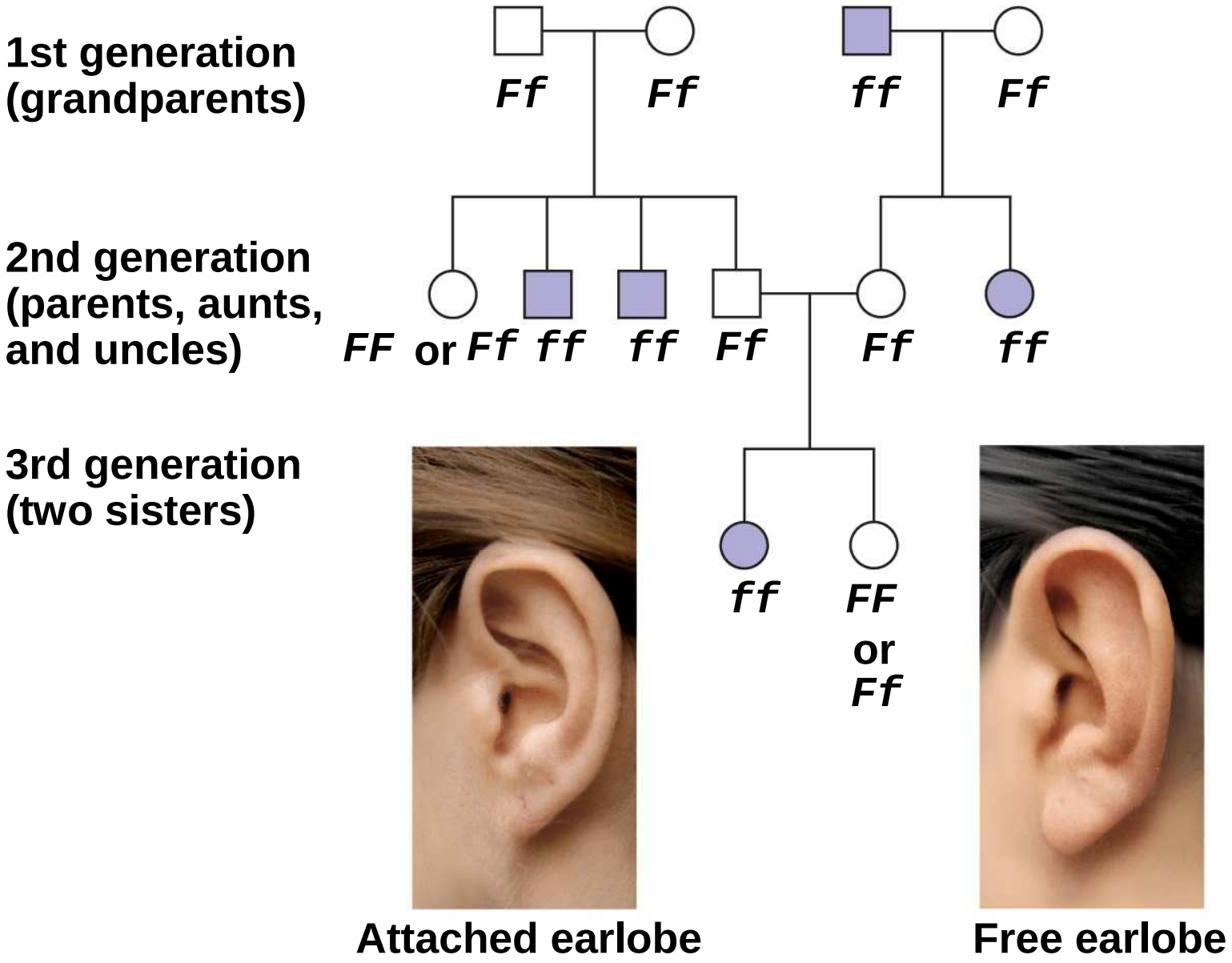


Fig. 14-15b



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

Fig. 14-15c



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

- Pedigrees can also be used to make predictions about future offspring
- We can use the multiplication and addition rules to predict the probability of specific phenotypes

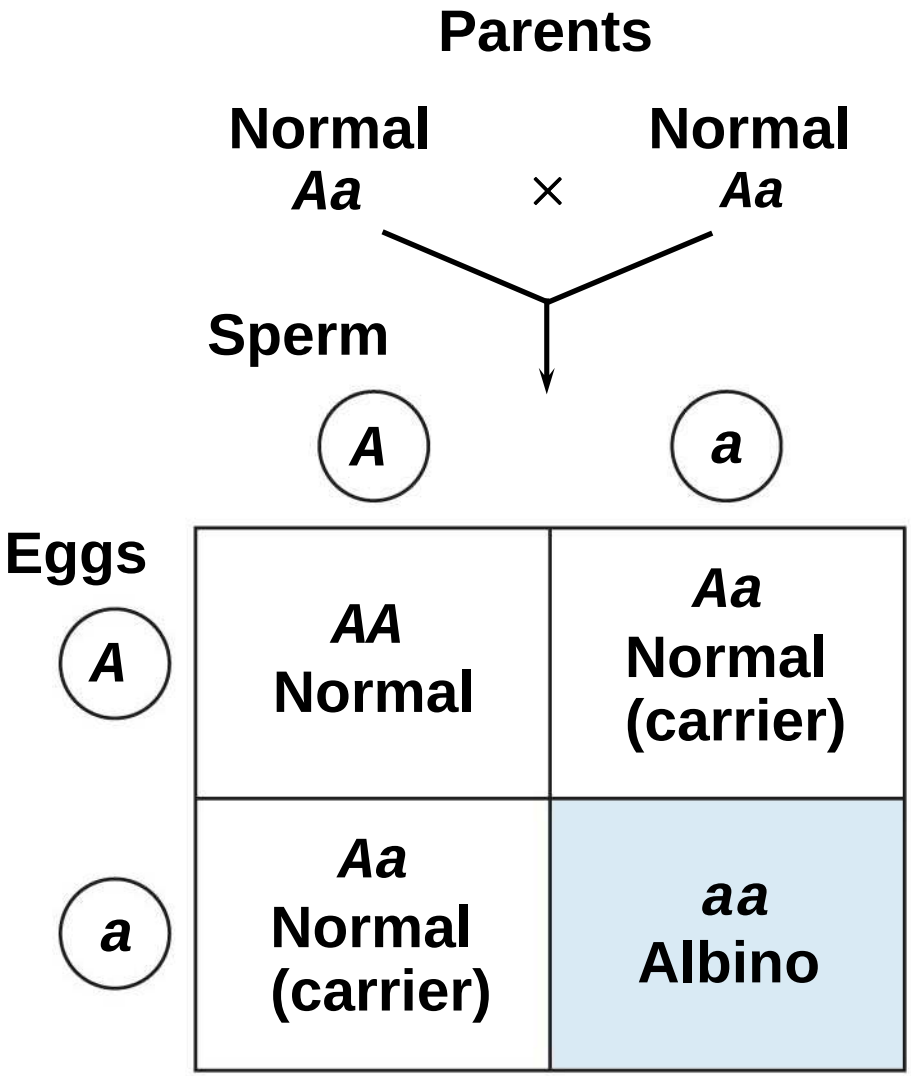
Recessively Inherited Disorders

- 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 and hair

Fig. 14-16



Why no consanguineous mating? [kon-**sang**-gwin-ee-*uh* s]

- 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

Cystic Fibrosis

- **Cystic fibrosis** is the most common lethal genetic disease in the United States,striking one out of every 2,500 people of European descent
- The cystic fibrosis allele results in defective or absent chloride transport channels in plasma membranes
- Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine

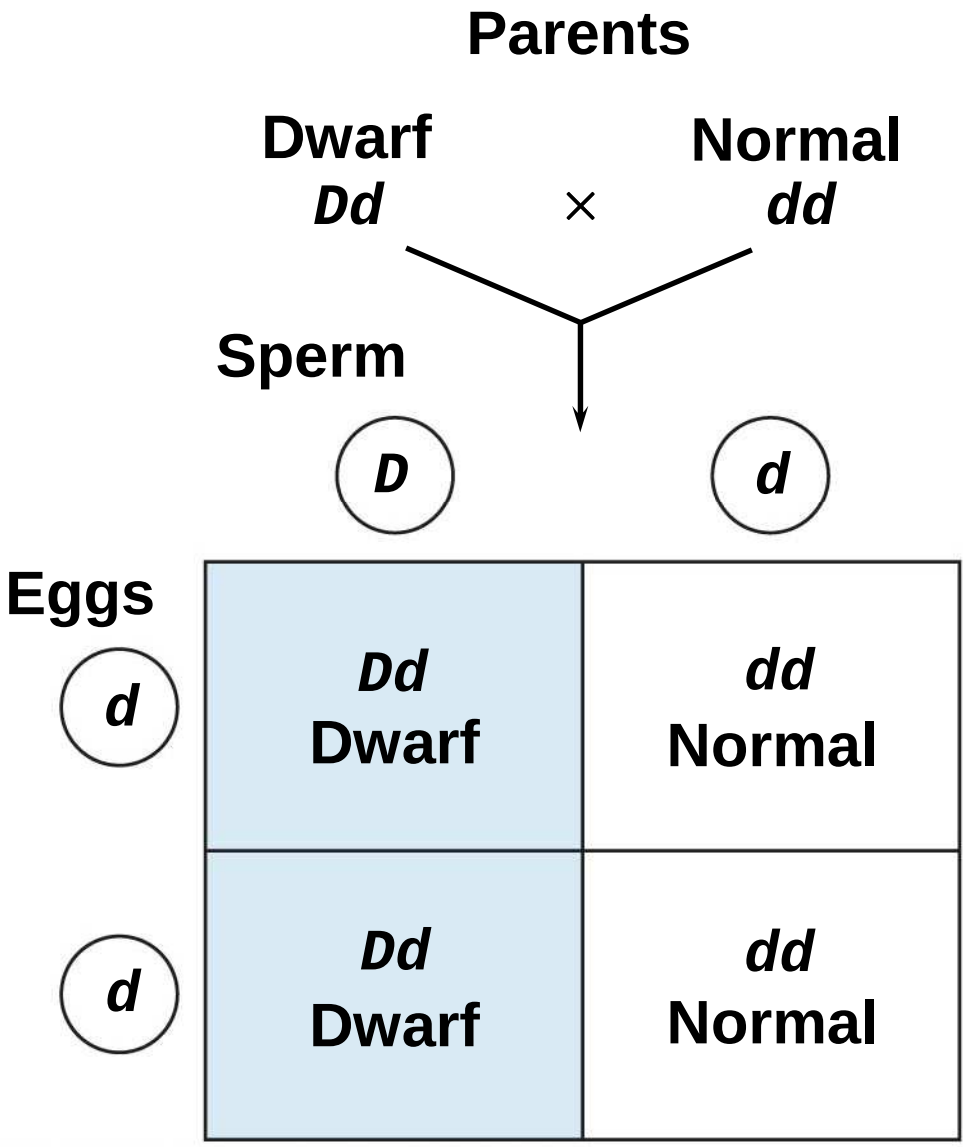
Sickle-Cell Disease

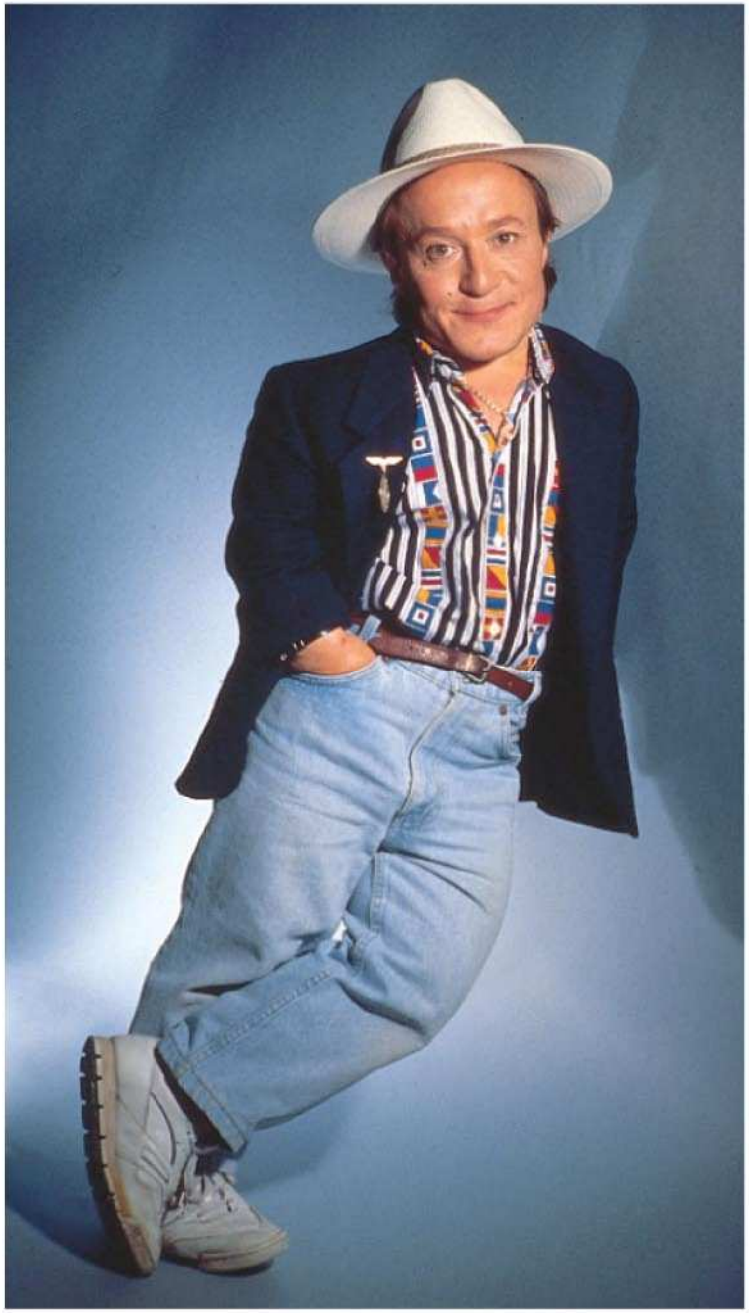
- **Sickle-cell disease** affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- Symptoms include physical weakness, pain, organ damage, and even paralysis

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* [ey-kon-dr*uh*-**pley**-zh*uh*] is a form of dwarfism caused by a rare dominant allele

Fig. 14-17





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

Genetic Testing and Counseling

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease

Counseling Based on Mendelian Genetics and Probability Rules

- Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders

Tests for Identifying Carriers

- For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately

Fetal Testing

- In **amniocentesis**, the liquid that bathes the fetus is removed and tested
- In **chorionic villus sampling (CVS)**, a sample of the placenta is removed and tested
- Other techniques, such as *ultrasound* and *fetoscopy*, allow fetal health to be assessed visually in utero

PLAY Video: Ultrasound of Human Fetus I

Fig. 14-18

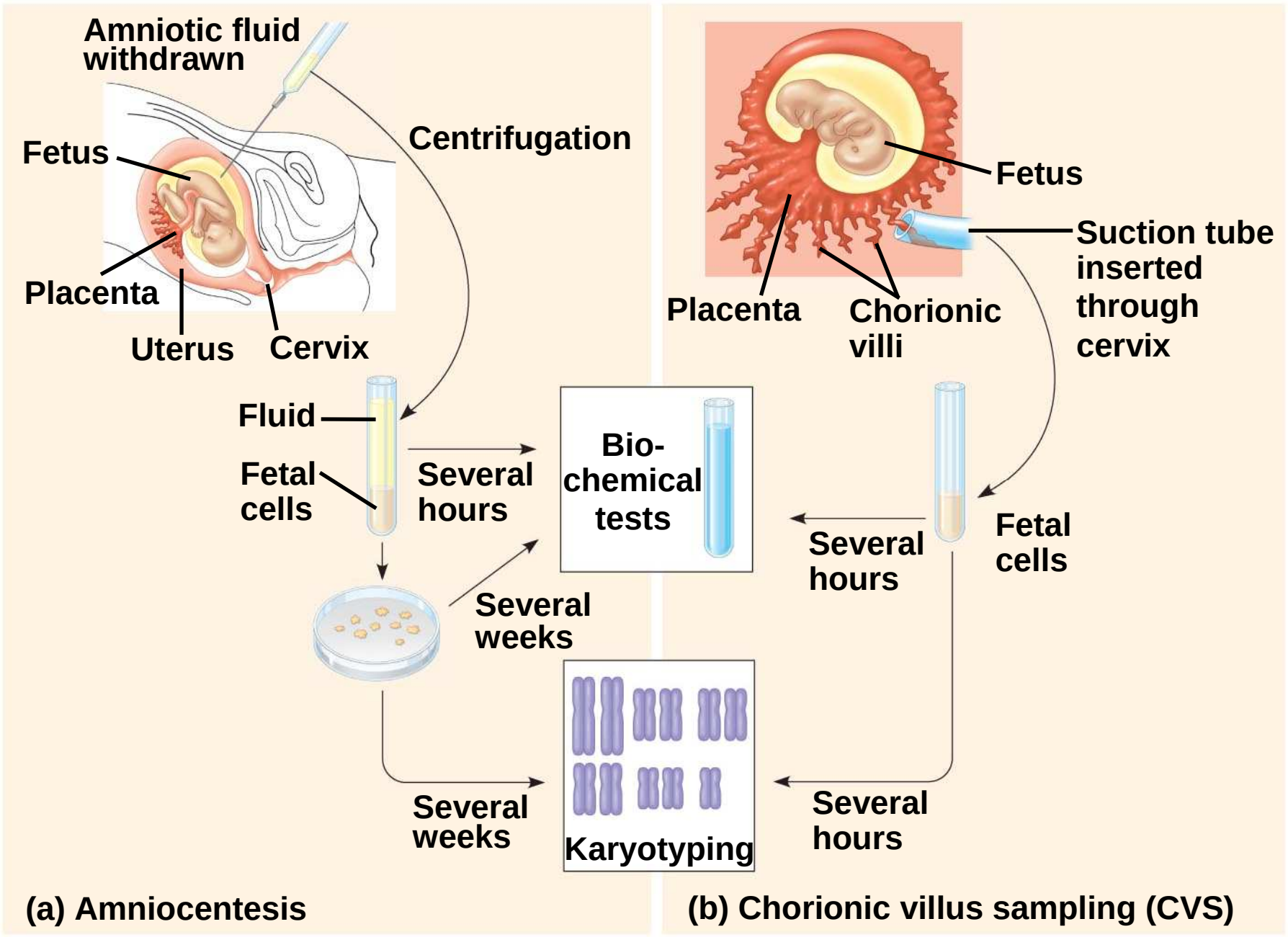


Fig. 14-18a

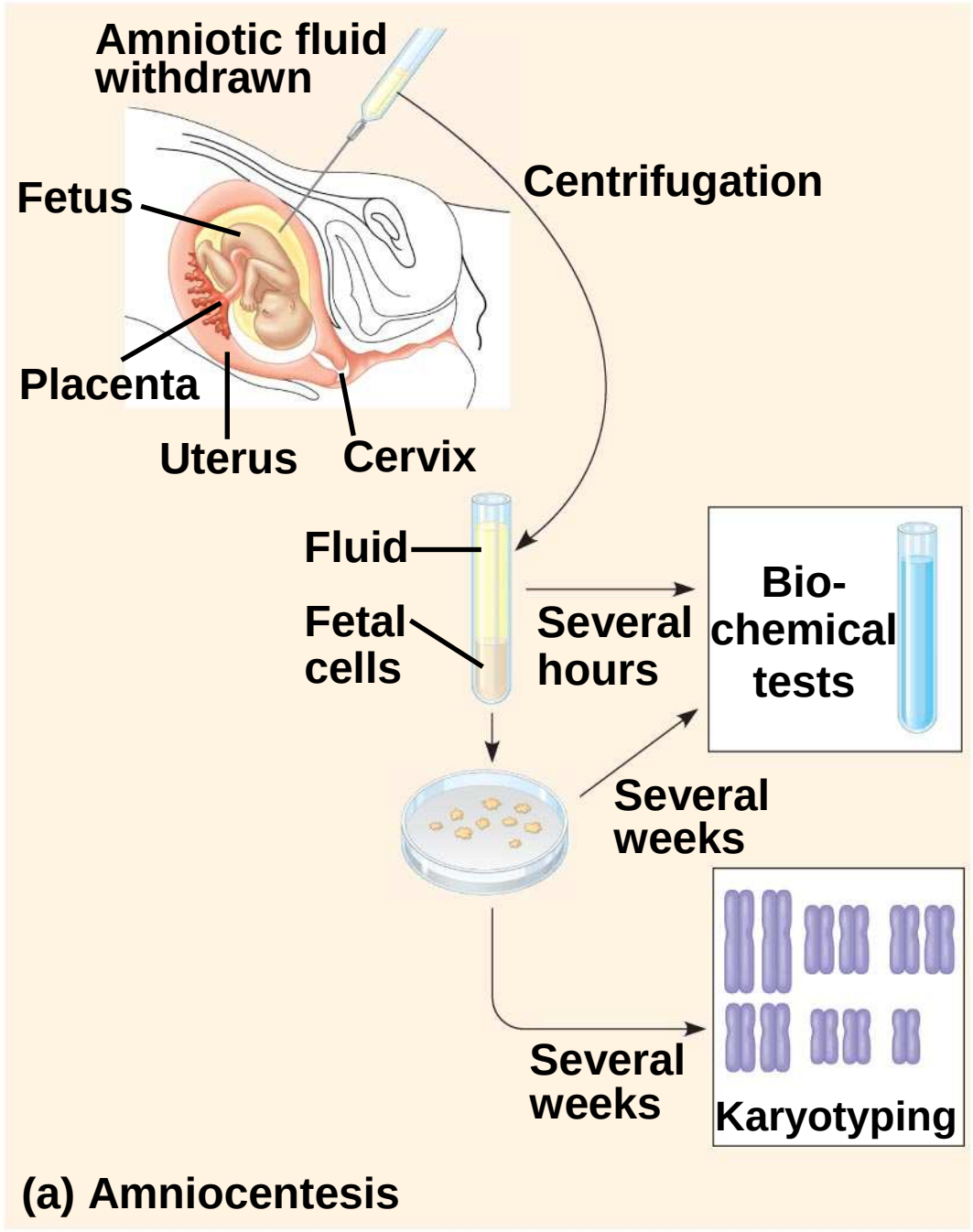
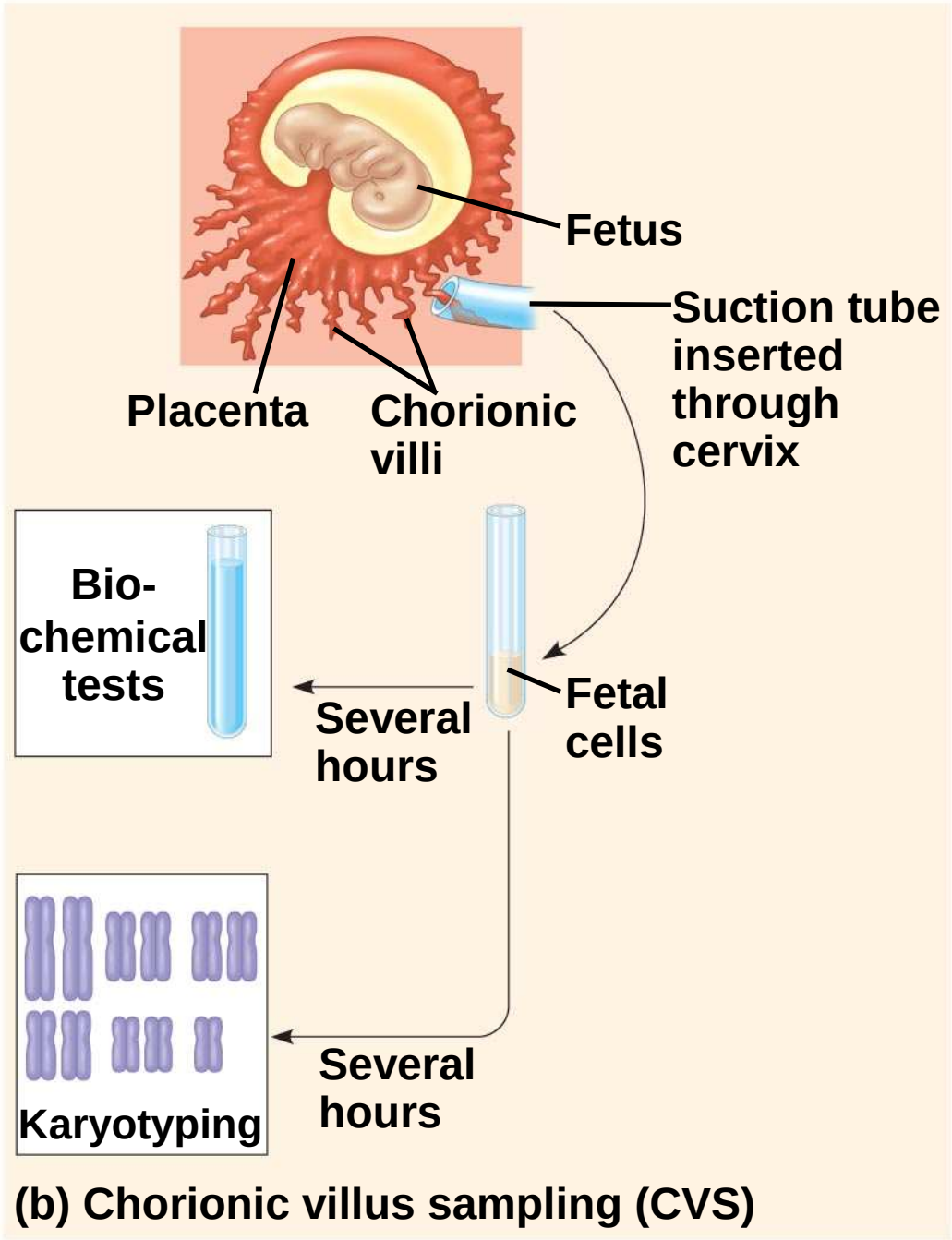


Fig. 14-18b



(b) Chorionic villus sampling (CVS)

Newborn Screening

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

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