Chapter 14

Mendel and the Gene Idea

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

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Overview: Drawing from the Deck of Genes

- What genetic principles account for the passing of traits from parents to offspring?
- The “blending” hypothesis is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)

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- The “particulate” hypothesis is the idea that parents pass on discrete heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas

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Fig. 14-1

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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

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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

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Fig. 14-2

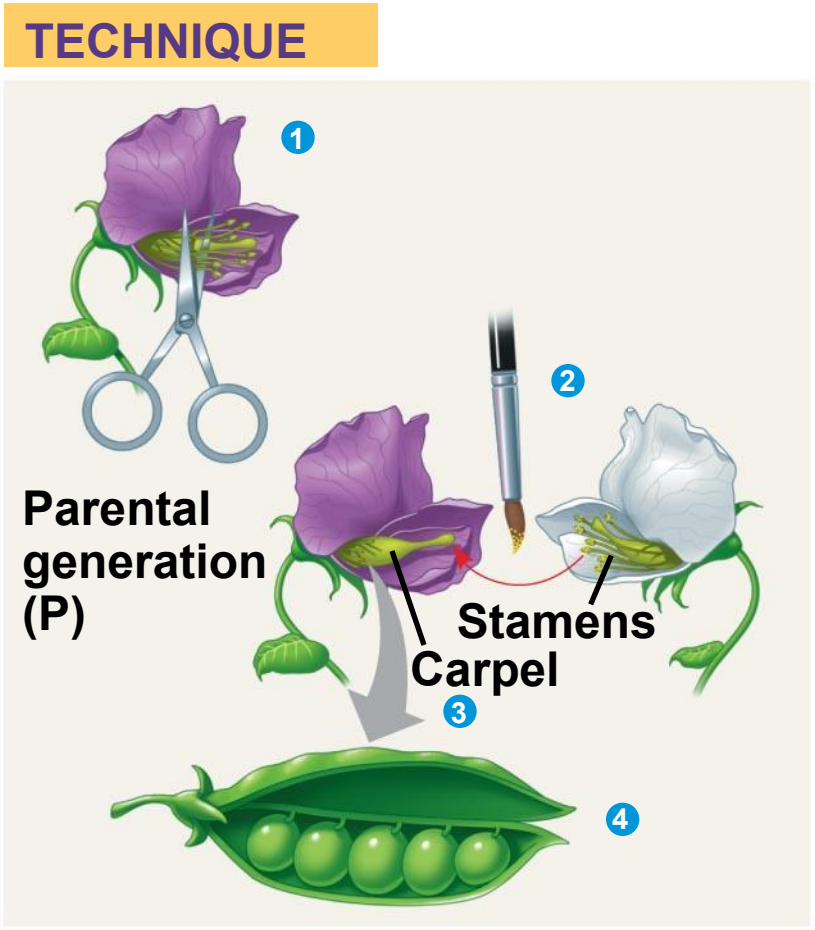
TECHNIQUE
1
2
Parental generation (P)
Stamens
Carpel
3
4
RESULTS
First filial gener-ation offsprin g (F1)
5
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Fig. 14-2a

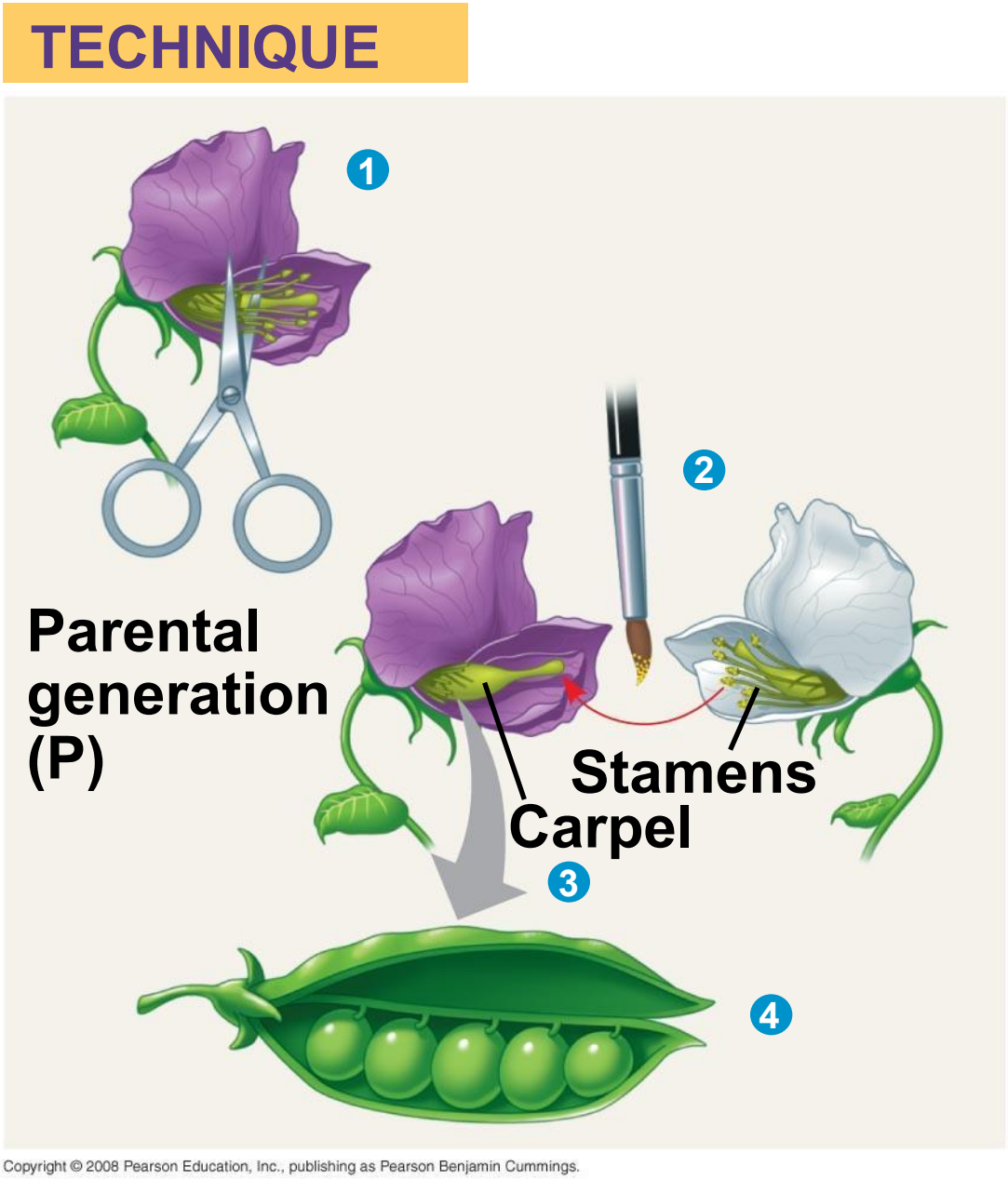

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Fig. 14-2b

RESULTS

First filial generation offspring (F_1)

5

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

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- 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

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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

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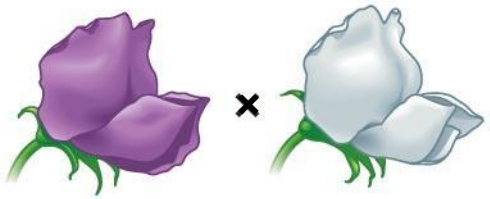
Fig. 14-3-1

EXPERIMENT

P Generation (true-breeding parents)



Purple flowers × **White flowers**

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Fig. 14-3-2

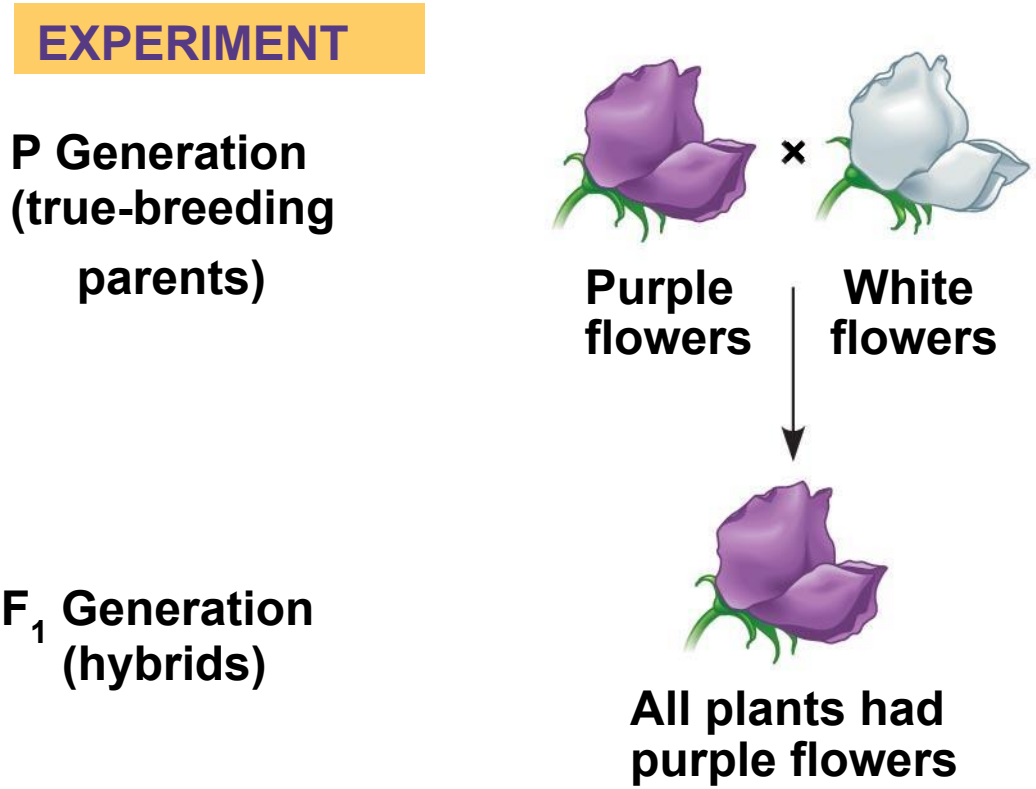

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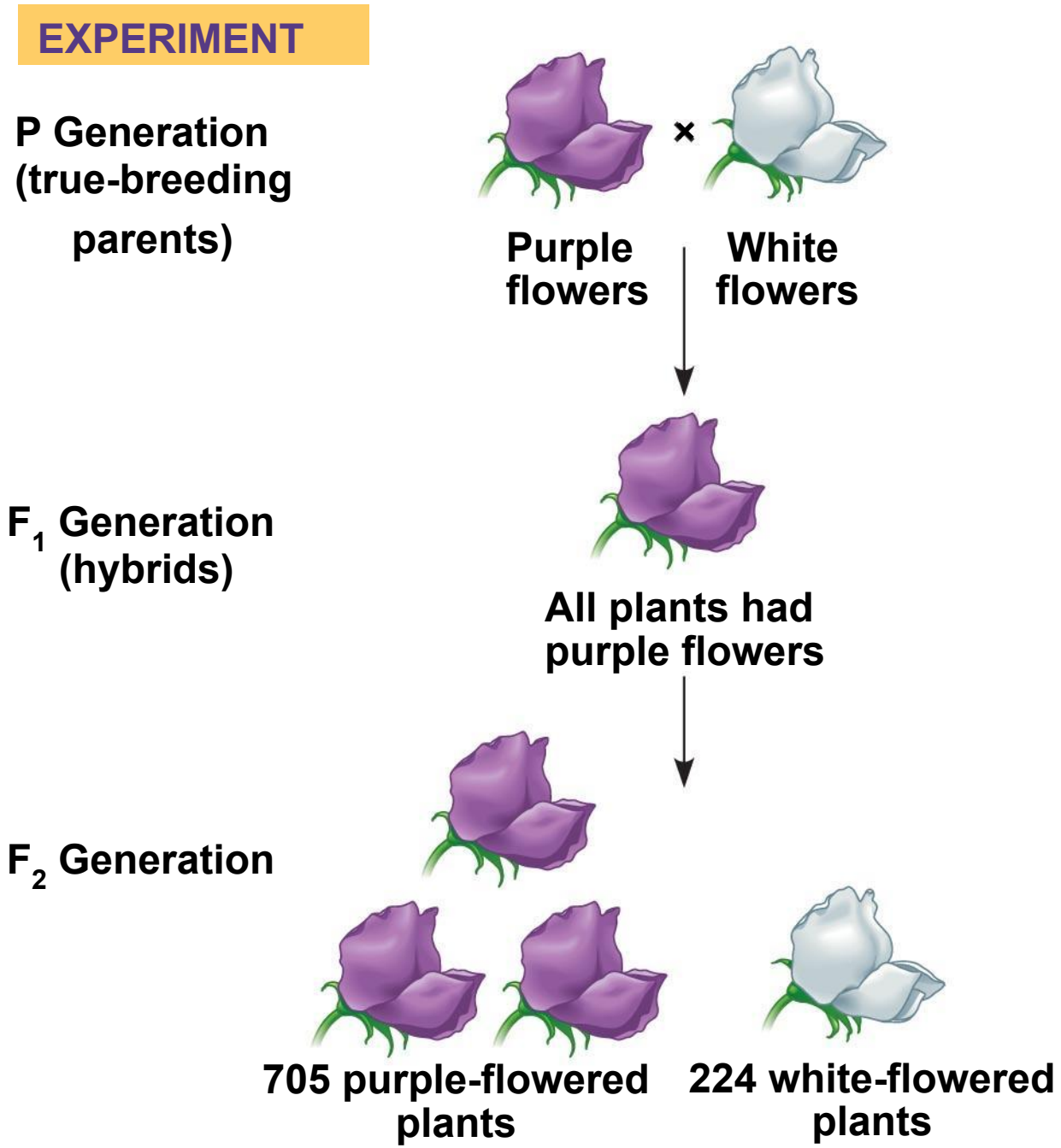
Fig. 14-3-3



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- 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

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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

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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
- These concepts can be related to what we now know about genes and chromosomes

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- 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

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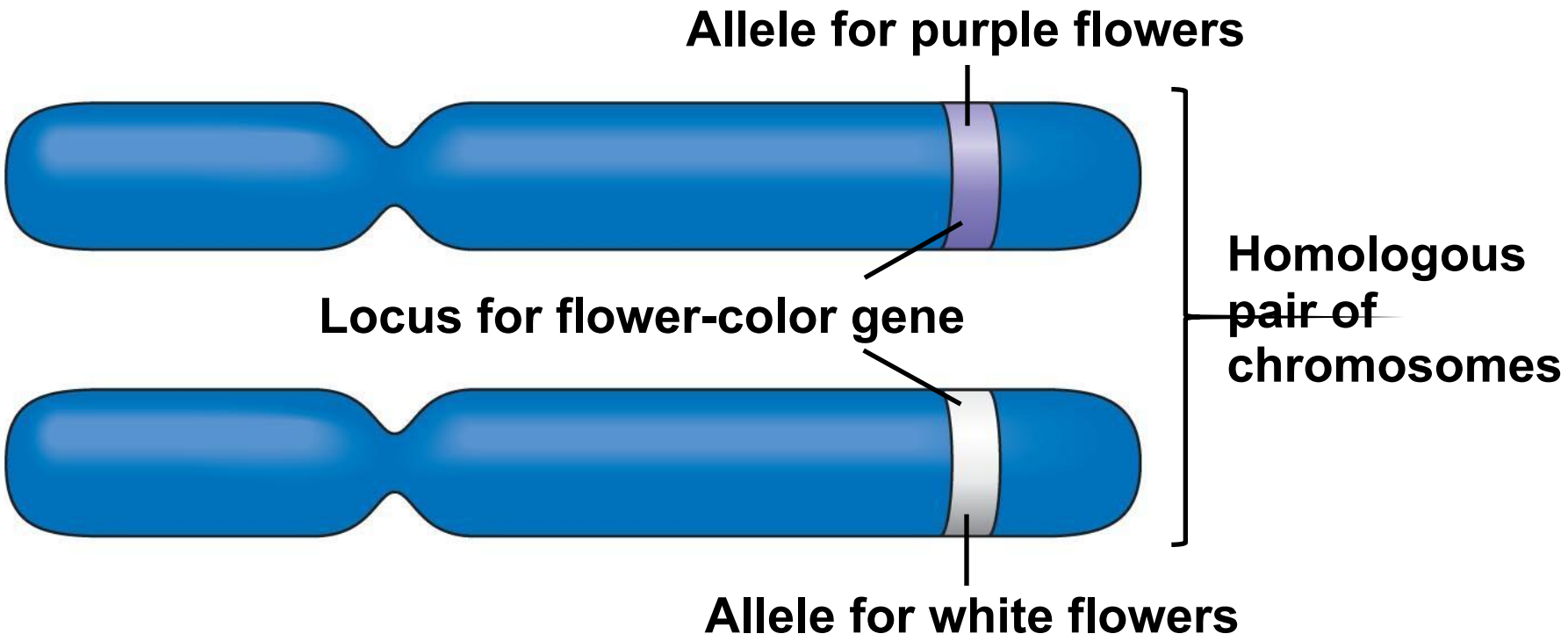
Fig. 14-4



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- 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

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- 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

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- 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

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- 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

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Fig. 14-5-1

P Generation

	Purple flowers	White flowers
Appearance:		
Genetic makeup:	*PP*	*p* *p*
Gametes:	*P*	*p*

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Fig. 14-5-2

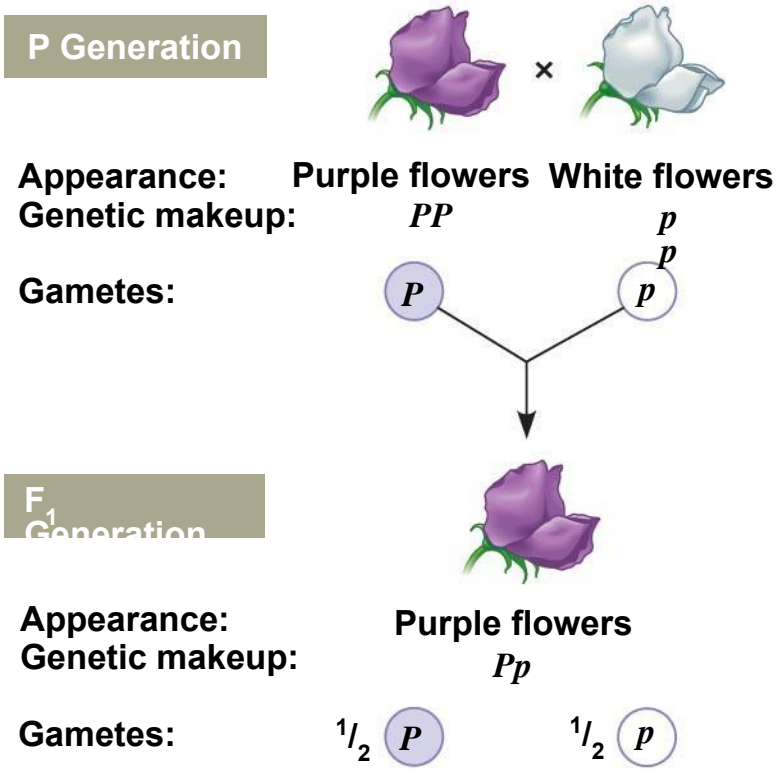

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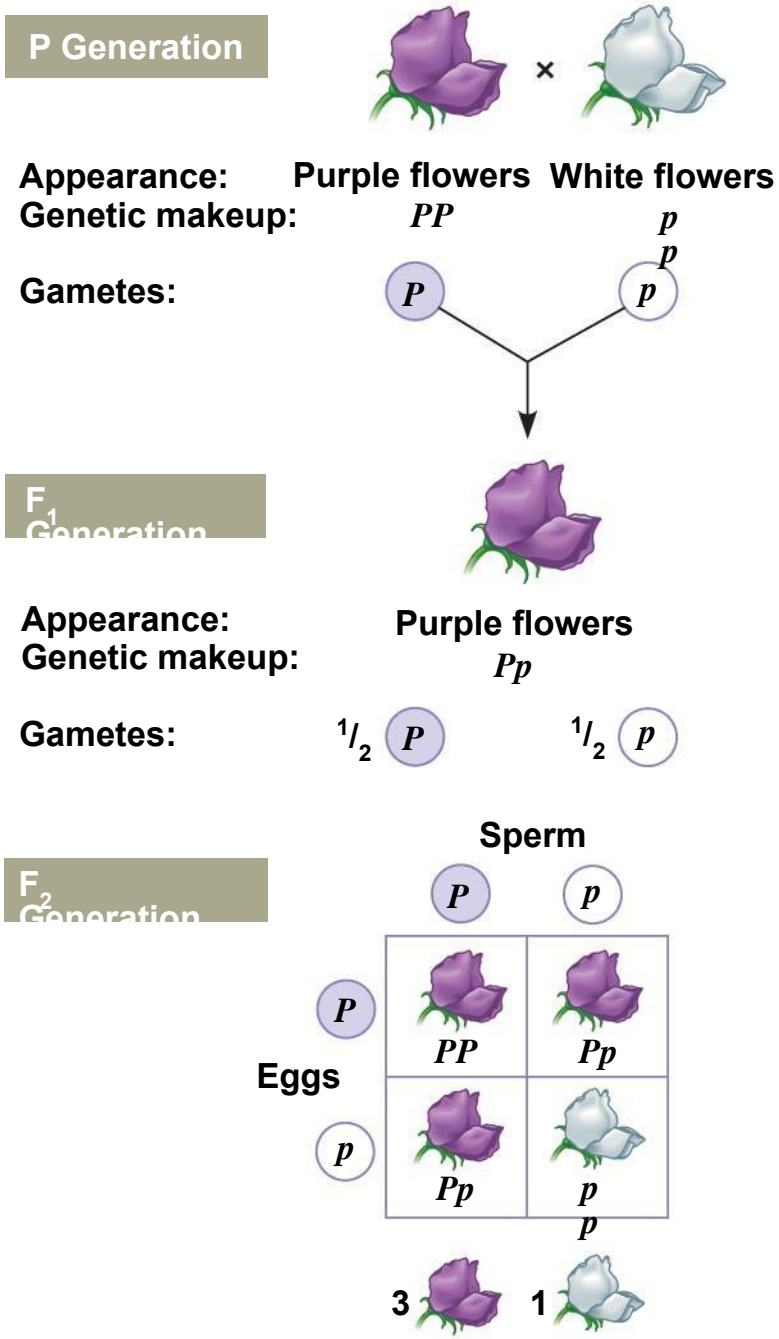
Fig. 14-5-3



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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

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- 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

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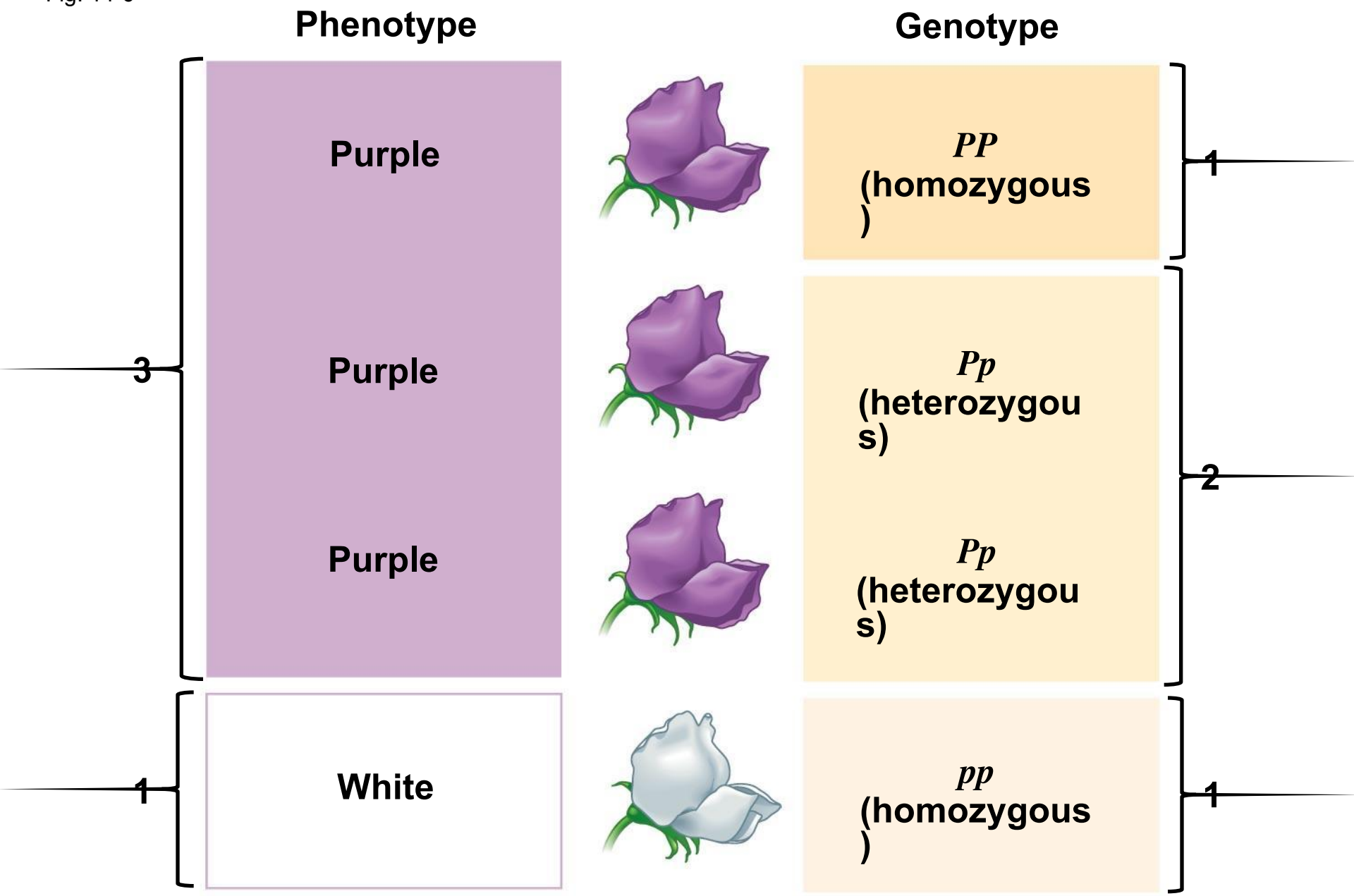
Fig. 14-6



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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

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Fig. 14-7

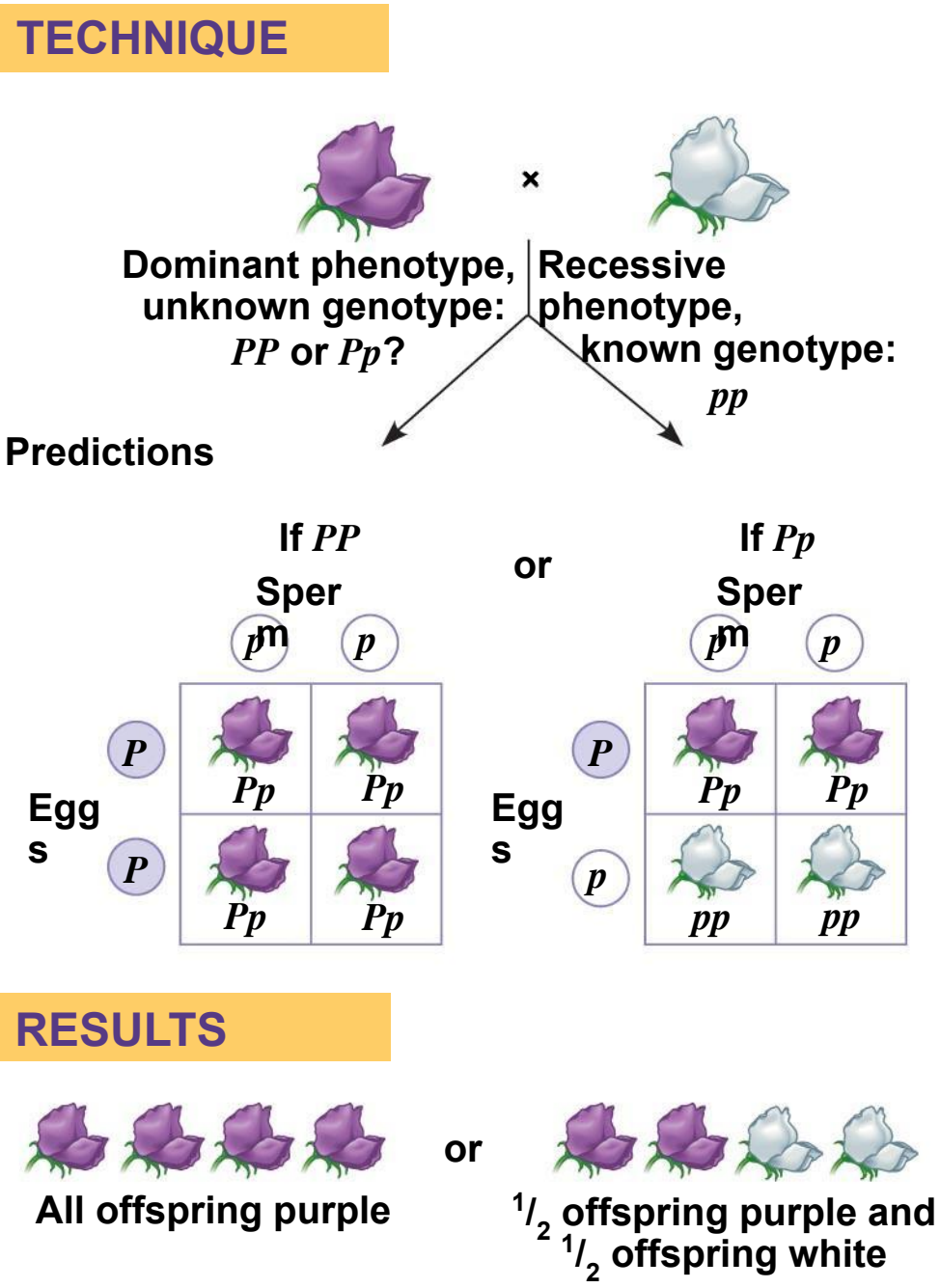

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Fig. 14-7a

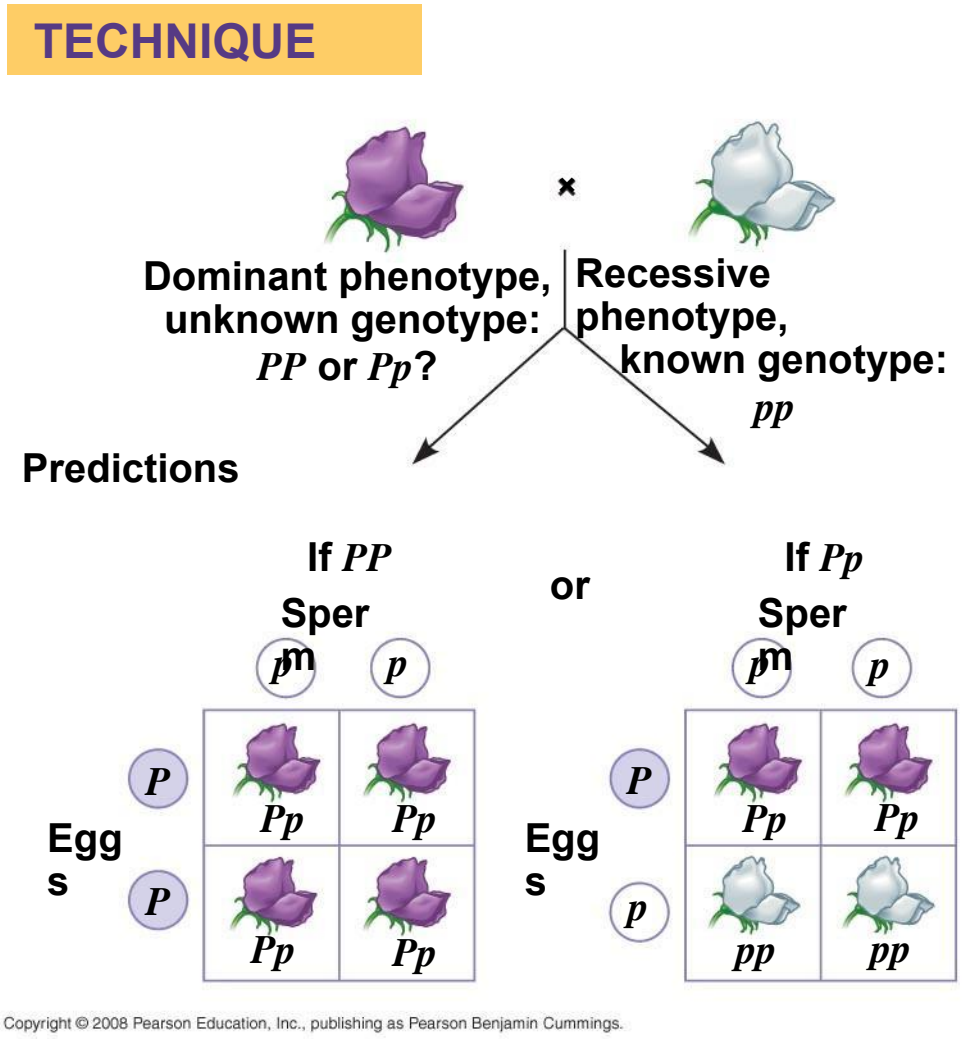

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Fig. 14-7b

RESULTS

All offspring purple

or

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

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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*

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- 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

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Fig. 14-8

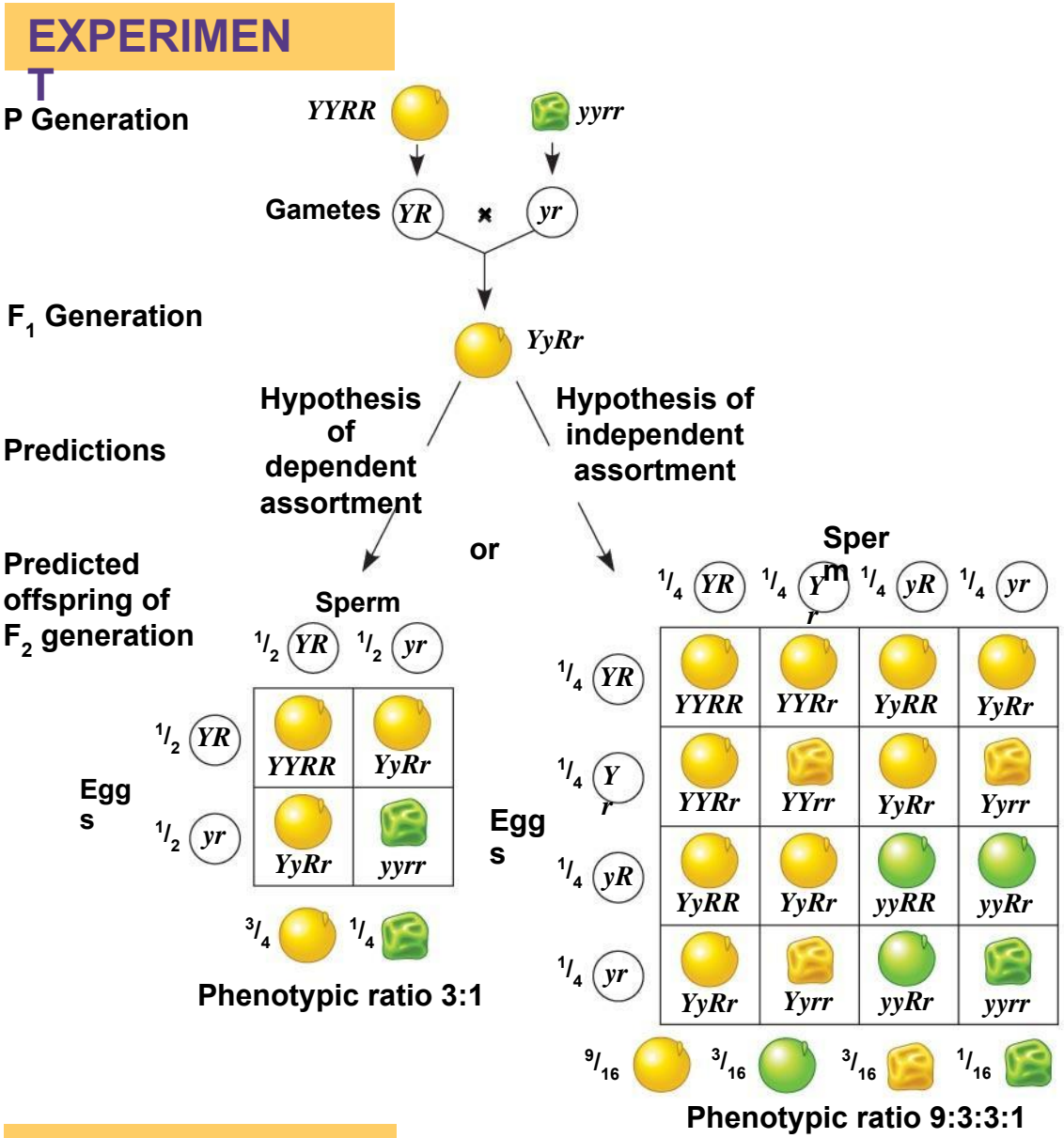

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Fig. 14-8a

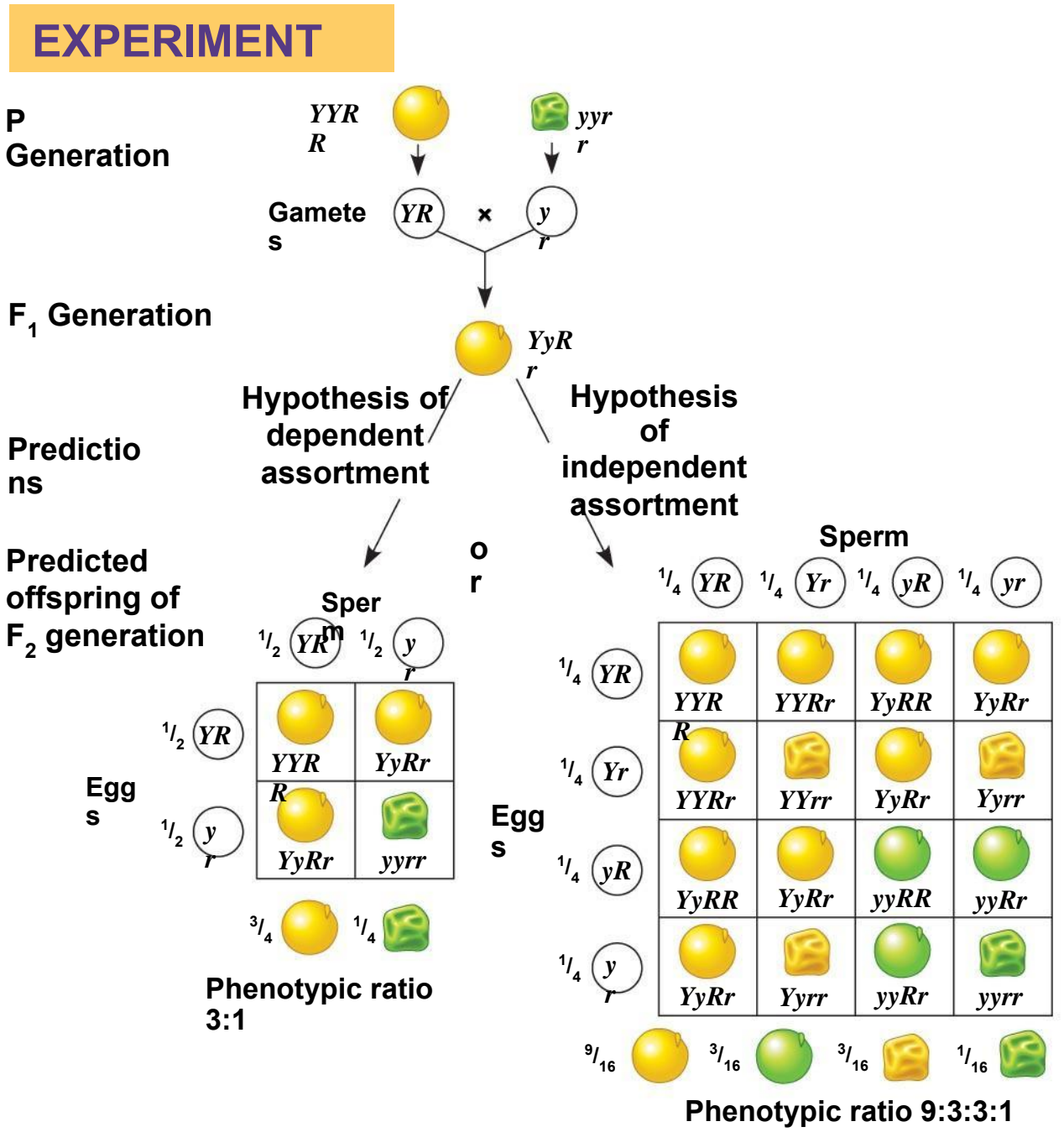

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Fig. 14-8b

RESULTS

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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

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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

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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 $\frac{1}{2}$ chance of carrying the dominant allele and a $\frac{1}{2}$ chance of carrying the recessive allele

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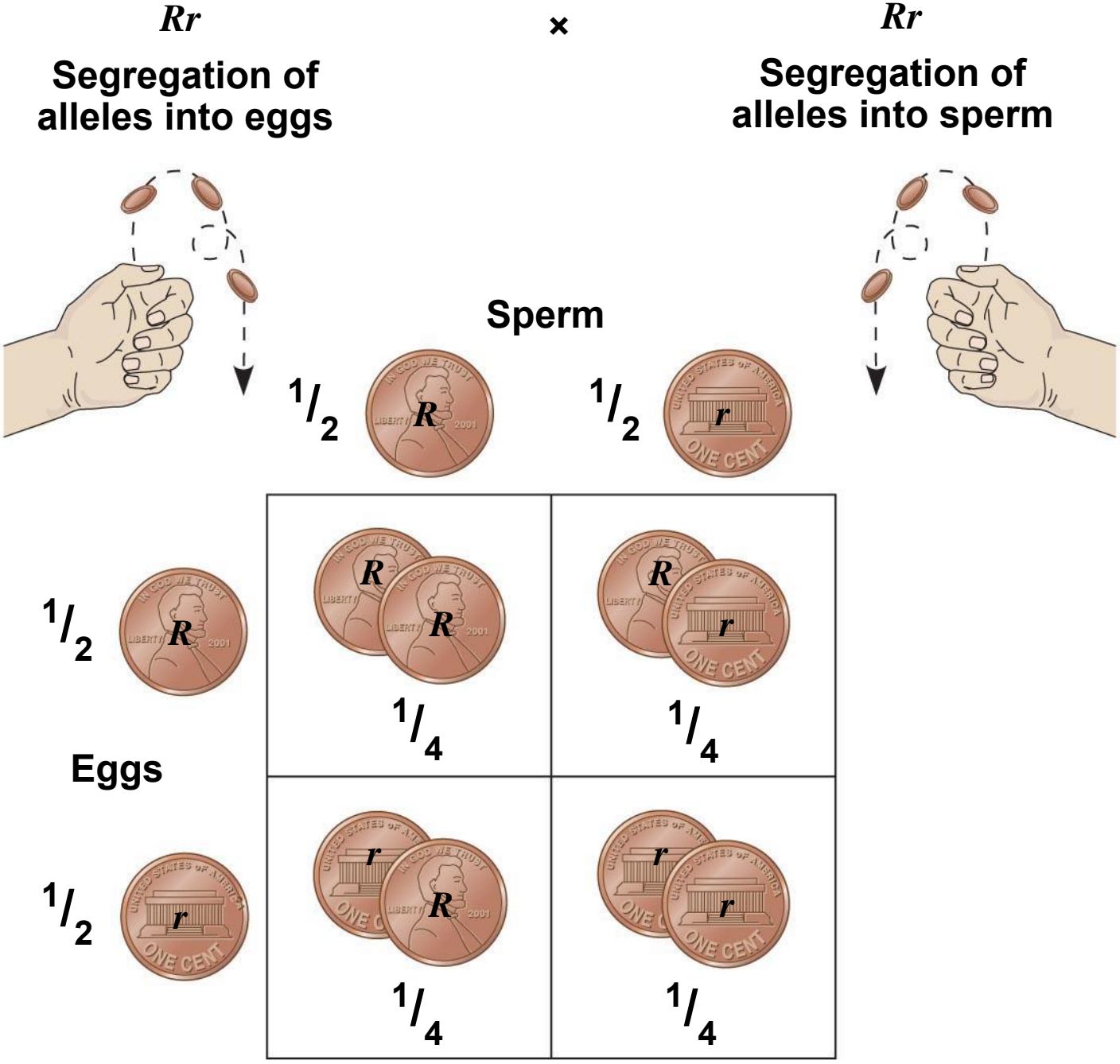
Fig. 14-9



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- 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

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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

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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**

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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

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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

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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

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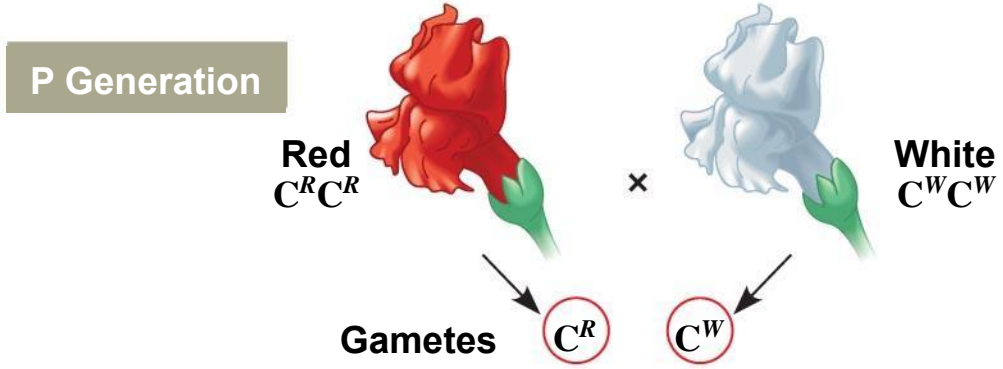
Fig. 14-10-1



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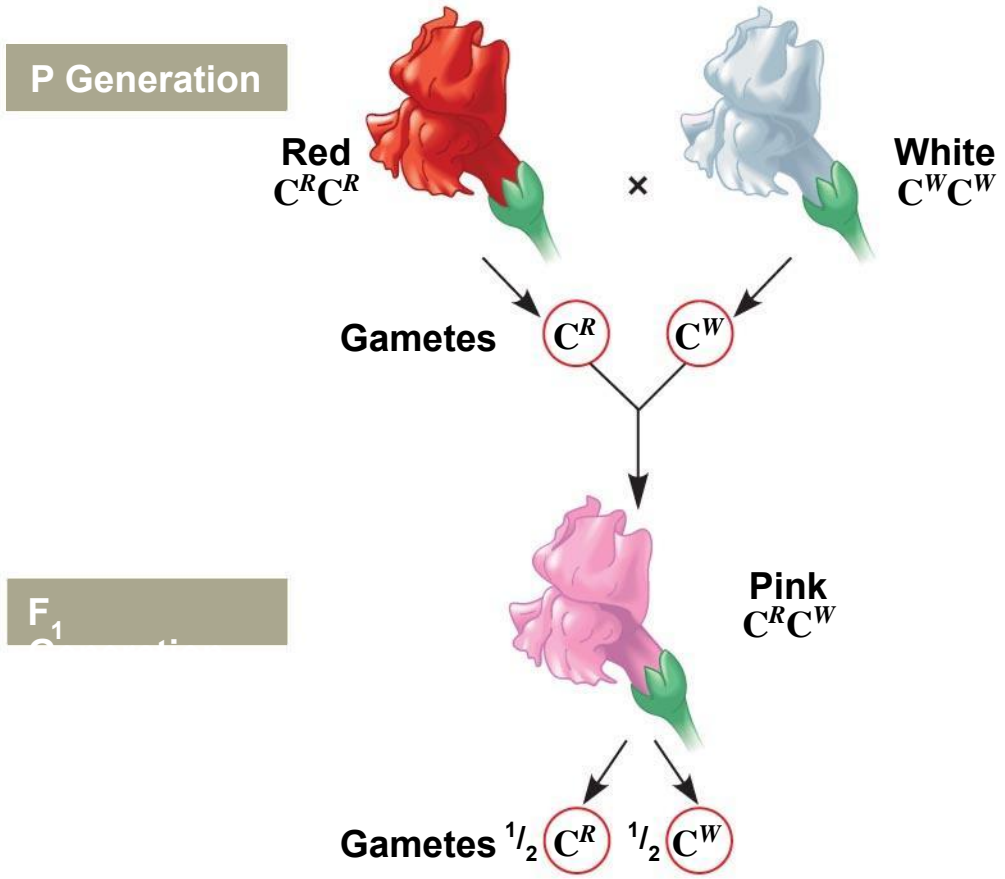

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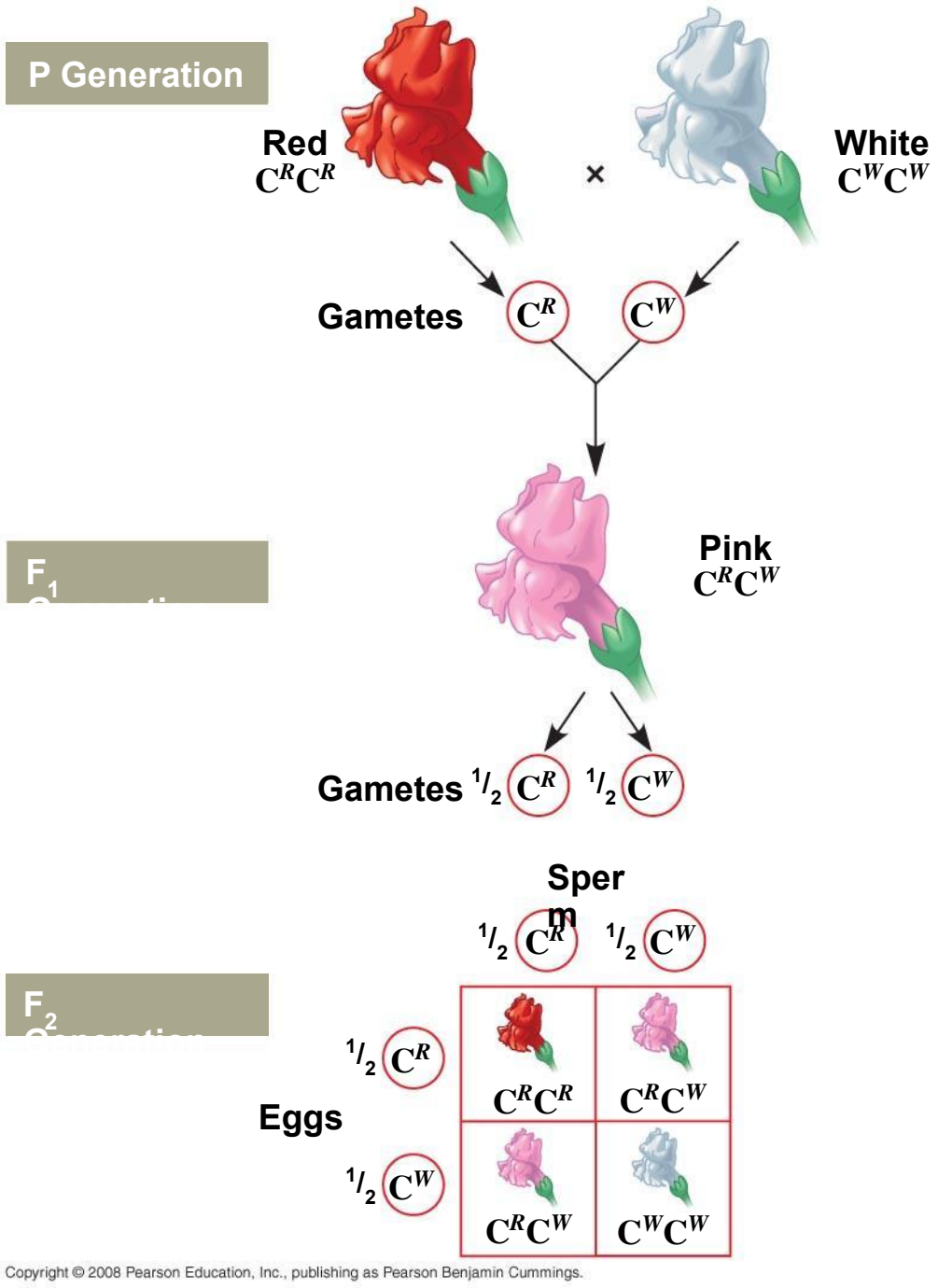

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The Relation Between Dominance and Phenotype

- 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

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- **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

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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

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- 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

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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

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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
$I^A I^B$		A B
ii		O

(b) Blood group genotypes and phenotypes

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Pleiotropy

- 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 and sickle-cell disease

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Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes

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Epistasis

- 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

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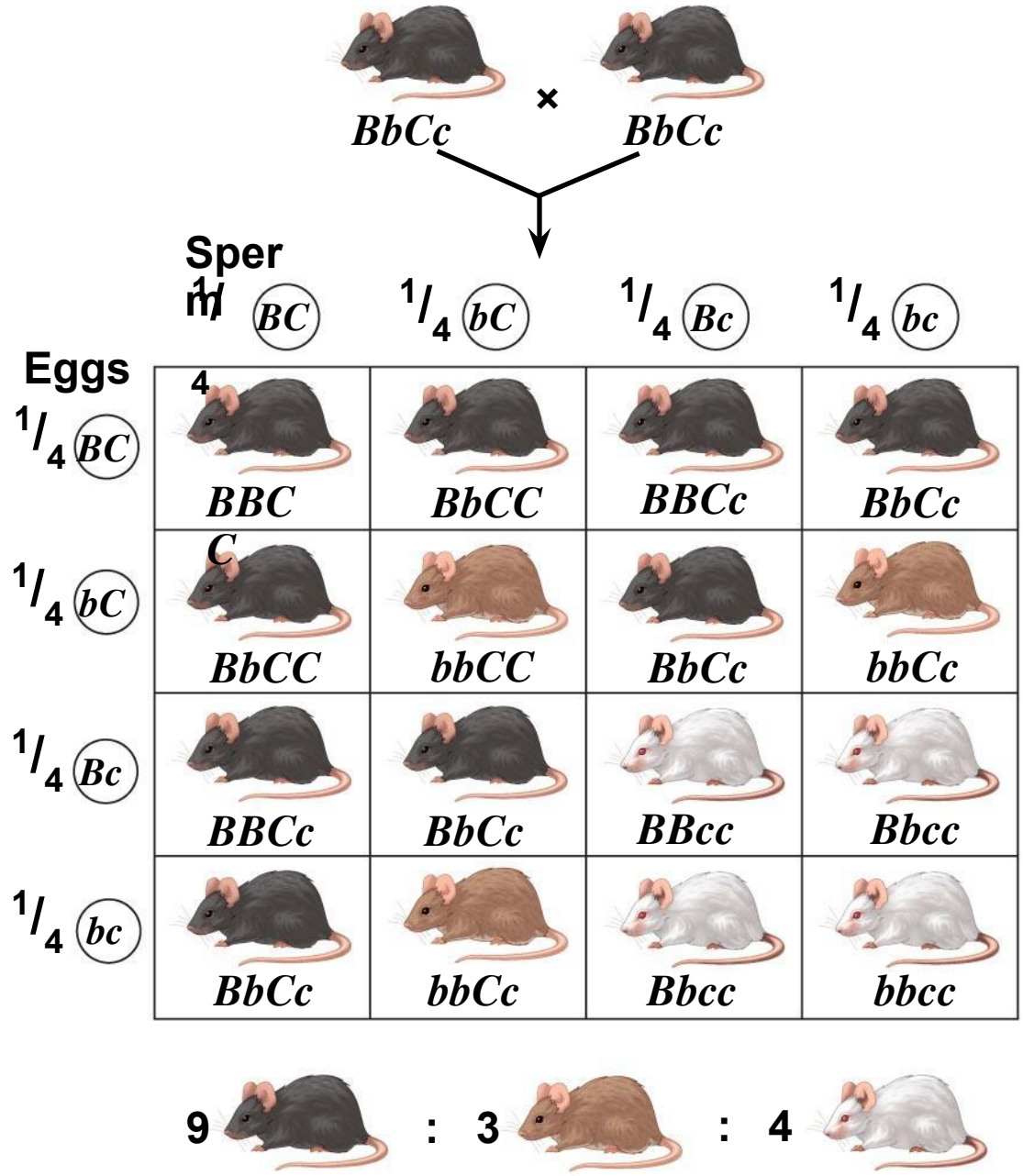
Fig. 14-12



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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

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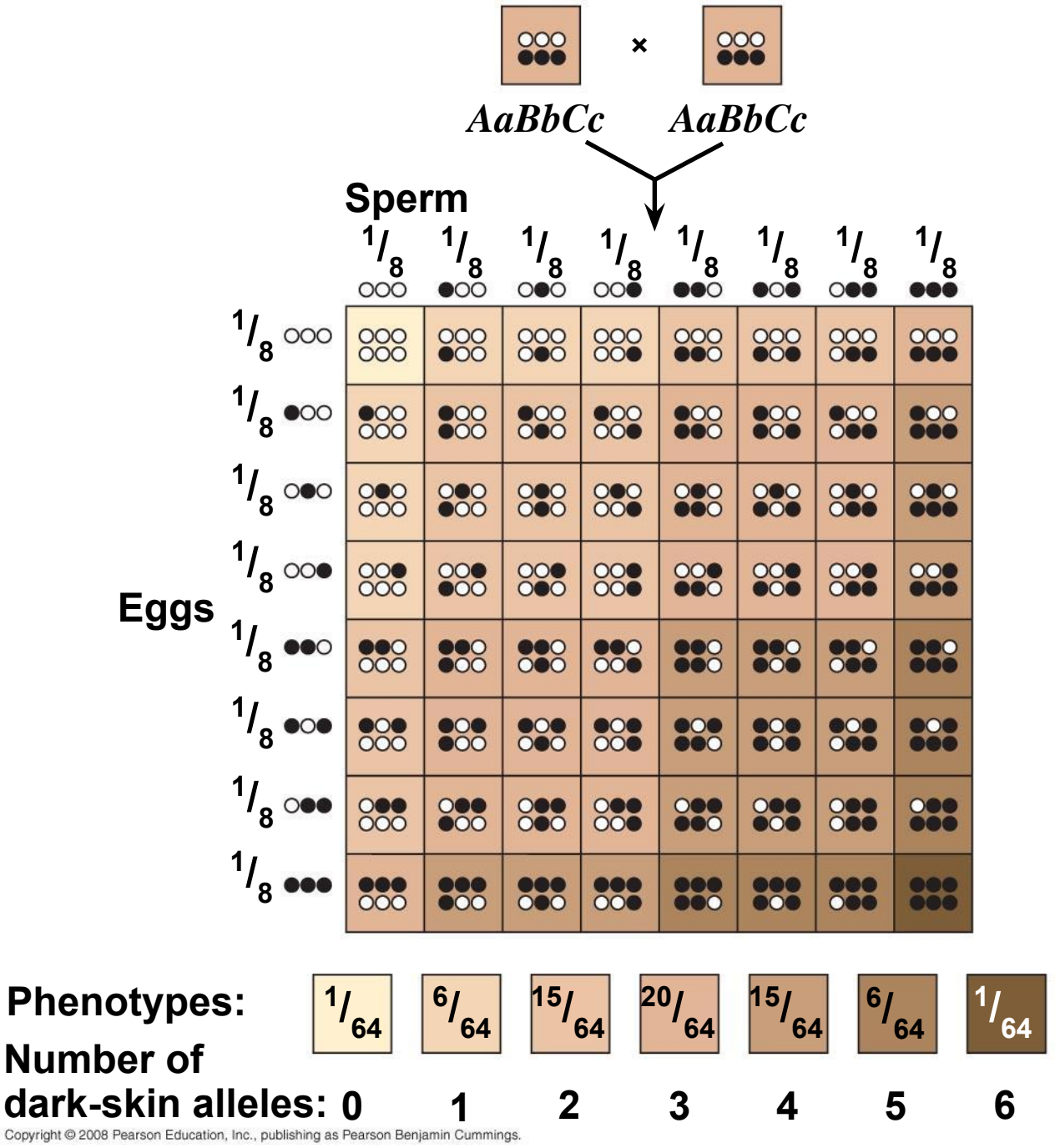
Fig. 14-13



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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

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Fig. 14-14

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

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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

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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

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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

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Fig. 14-15

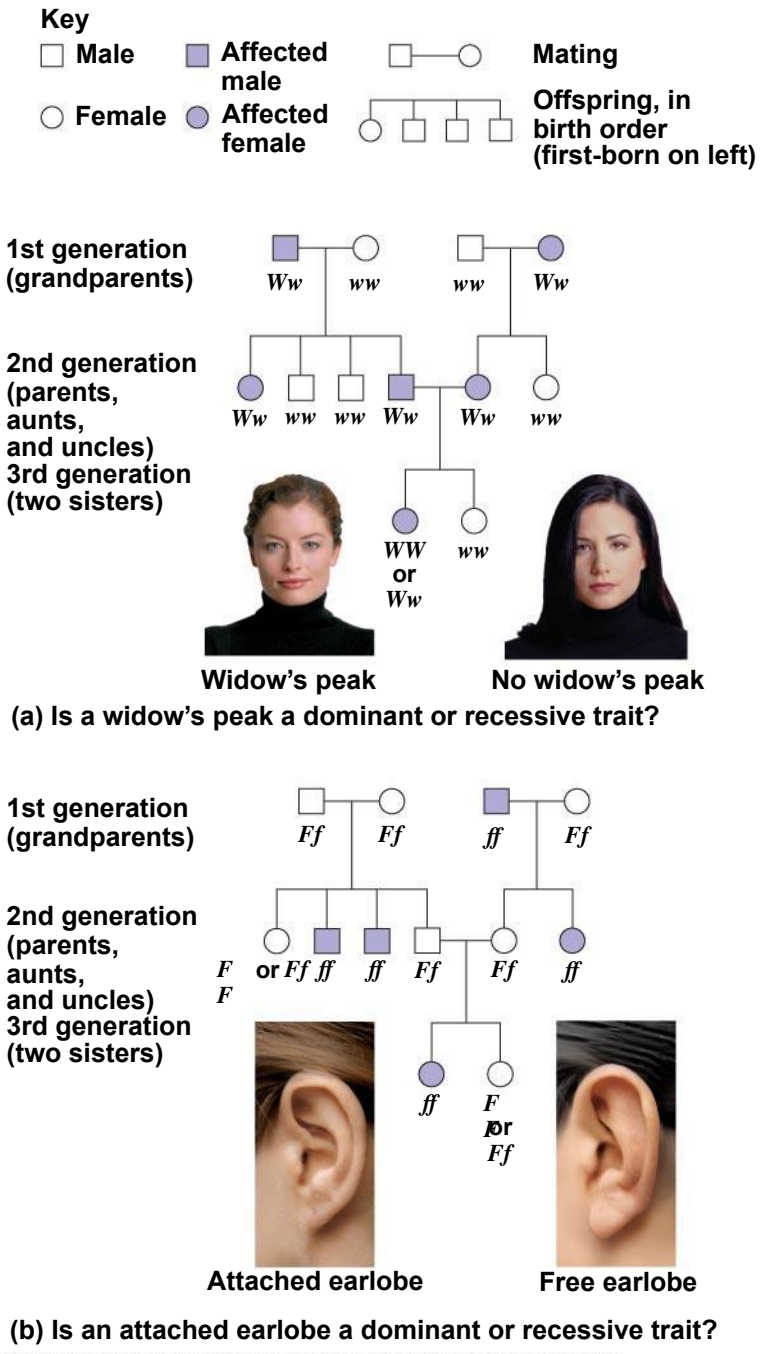

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Fig. 14-15a

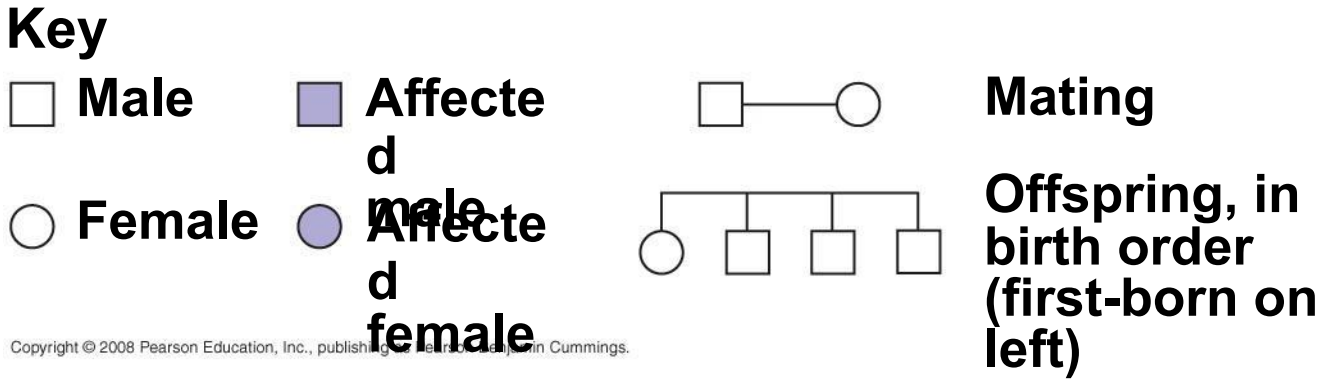

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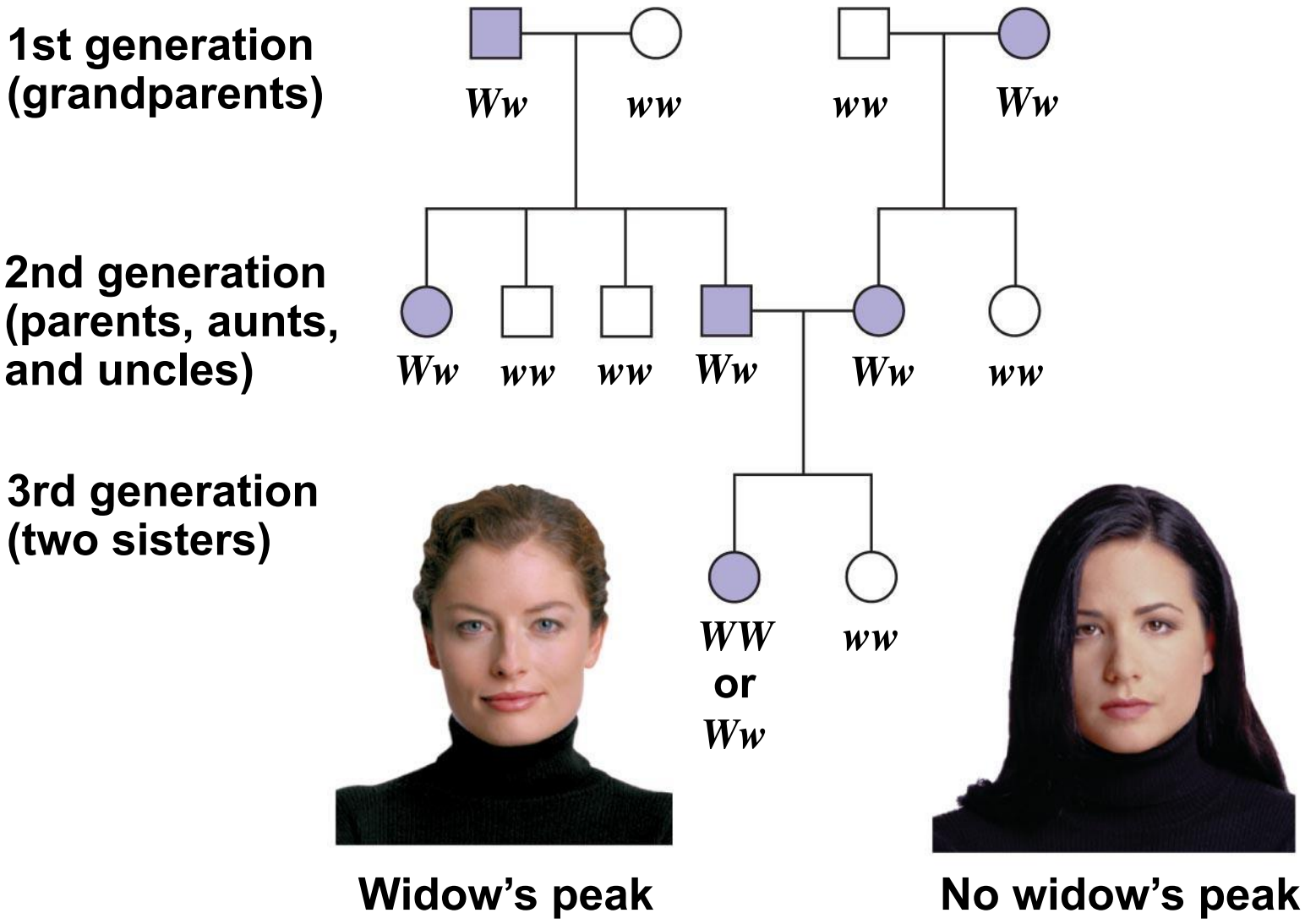
Fig. 14-15b



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

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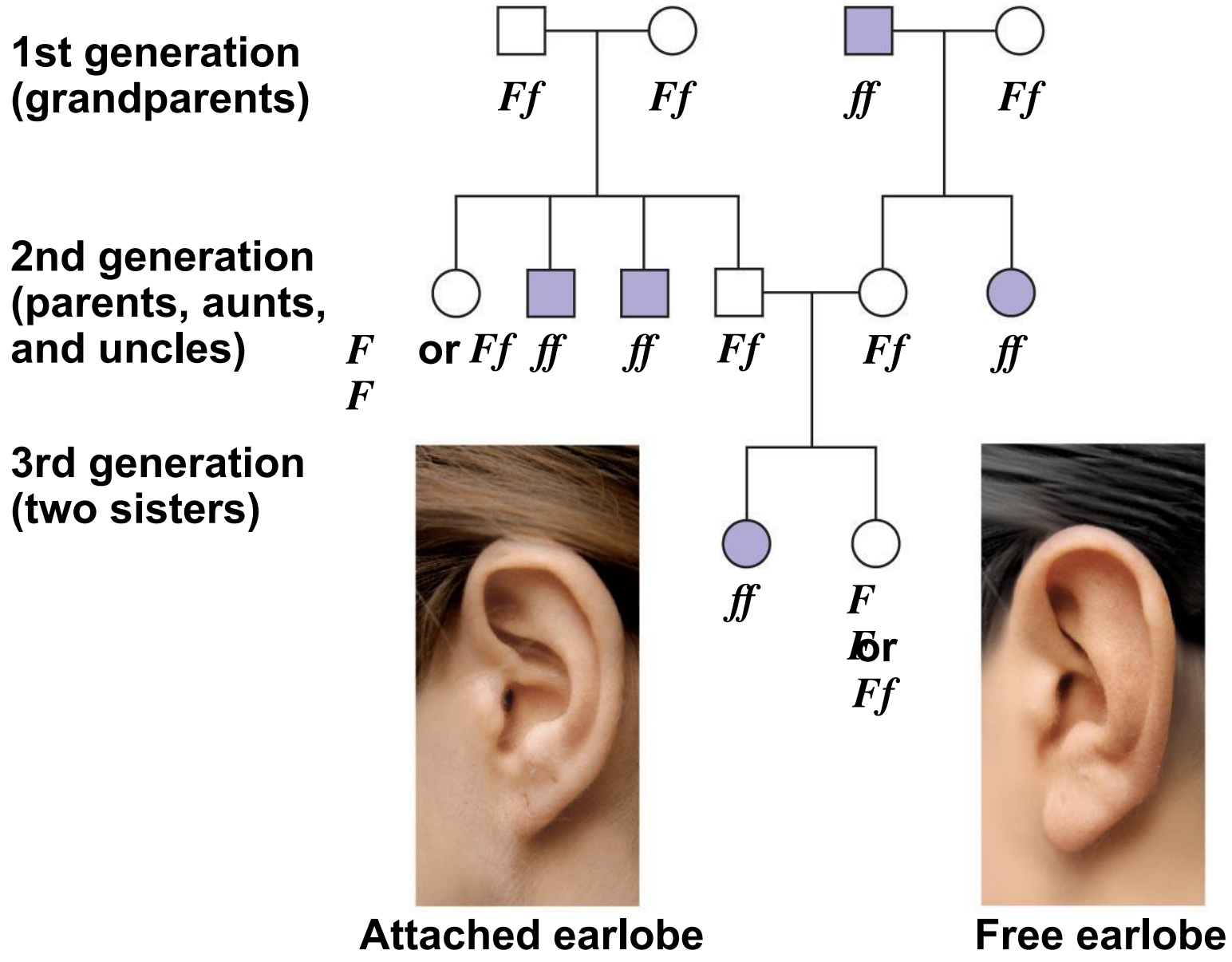
Fig. 14-15c



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

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- 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

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Recessively Inherited Disorders

- Many genetic disorders are inherited in a recessive manner

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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

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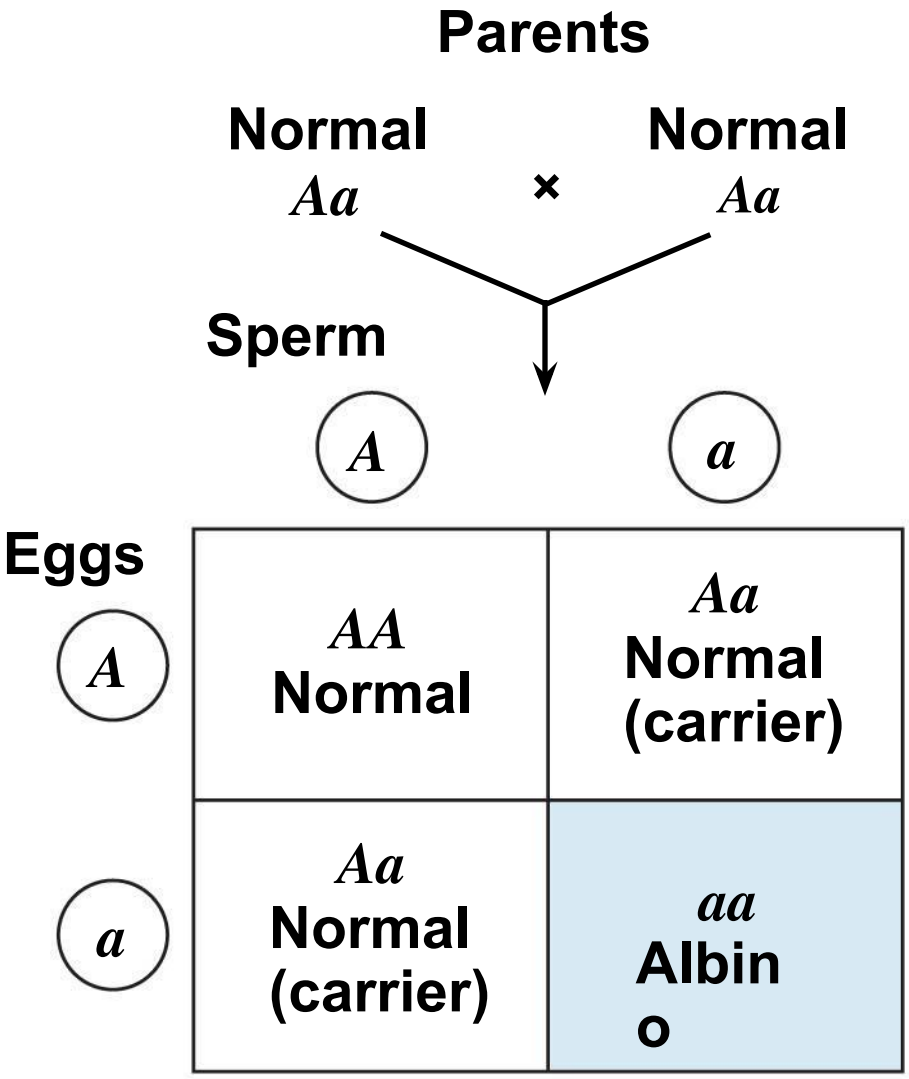
Fig. 14-16



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- 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

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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

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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

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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

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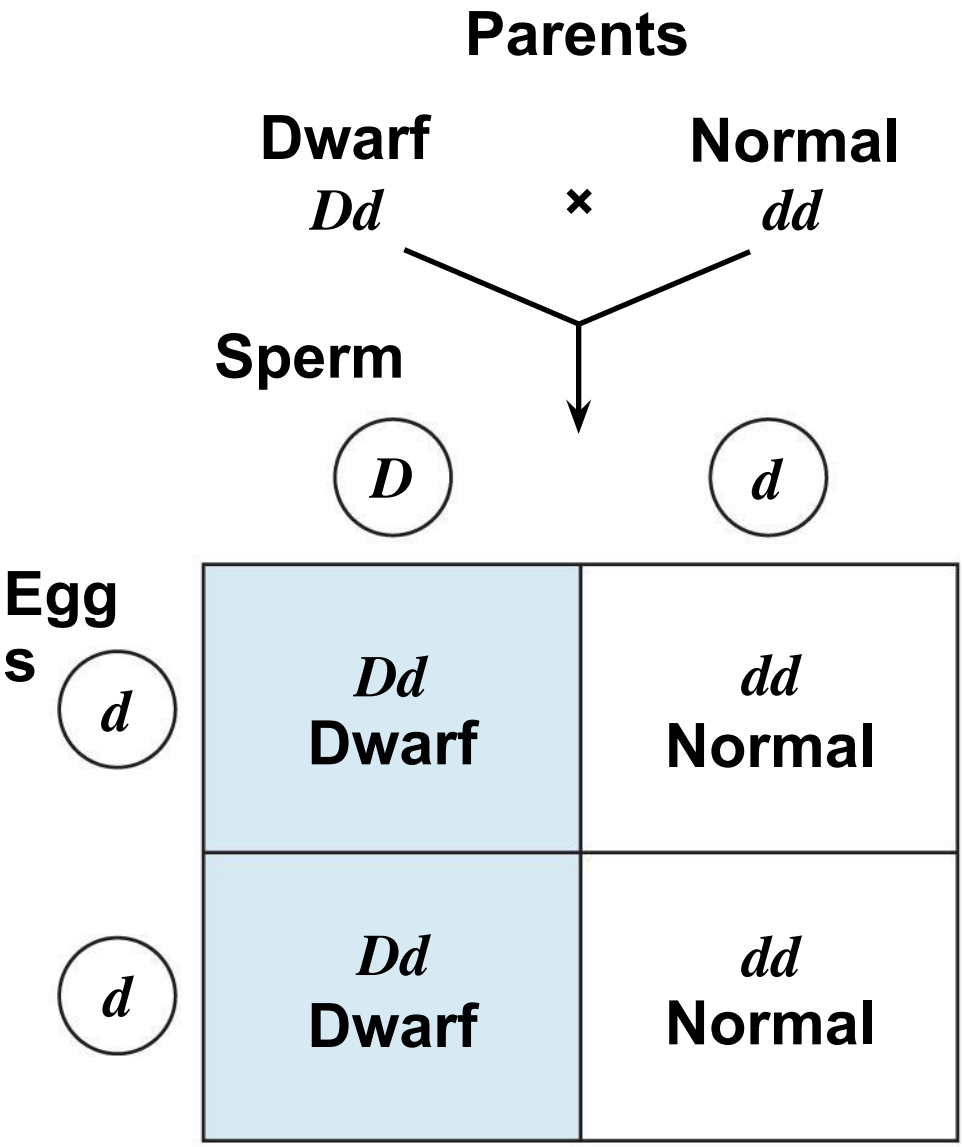
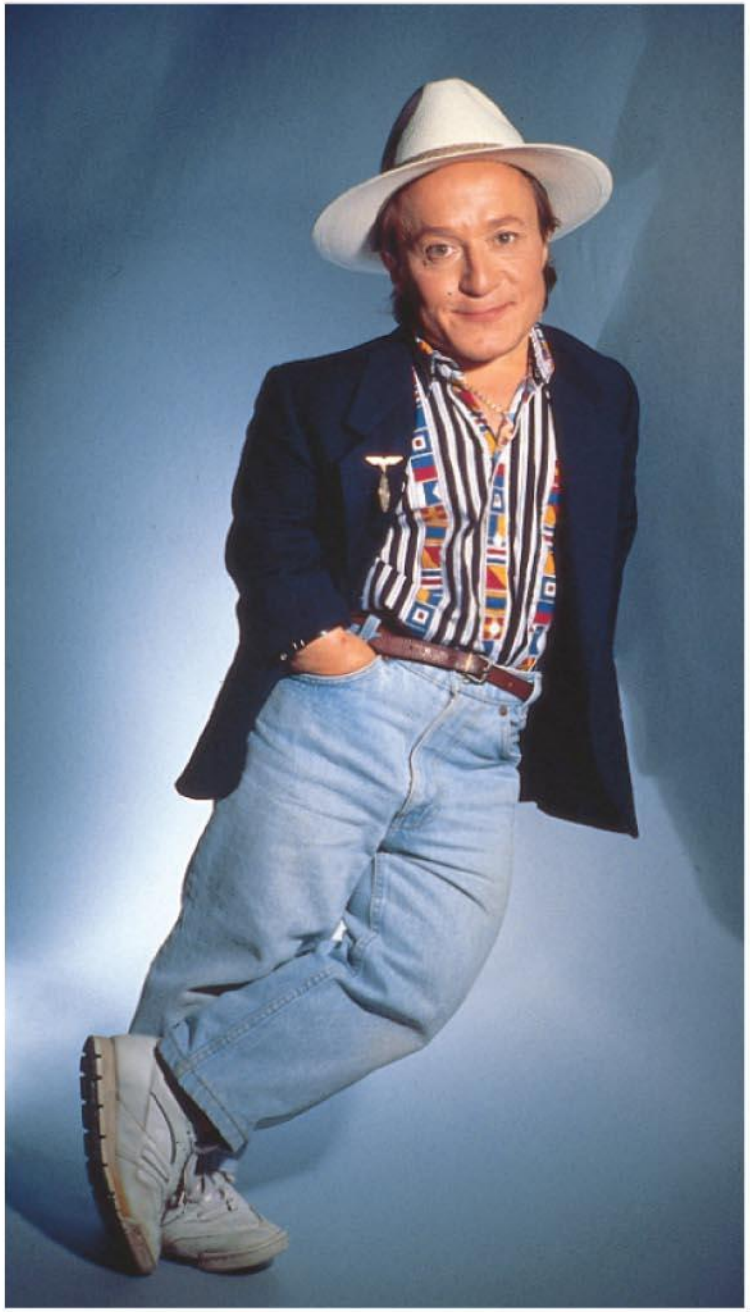
Fig. 14-17





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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

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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

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Genetic Testing and Counseling

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

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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

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Tests for Identifying Carriers

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

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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

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Fig. 14-18

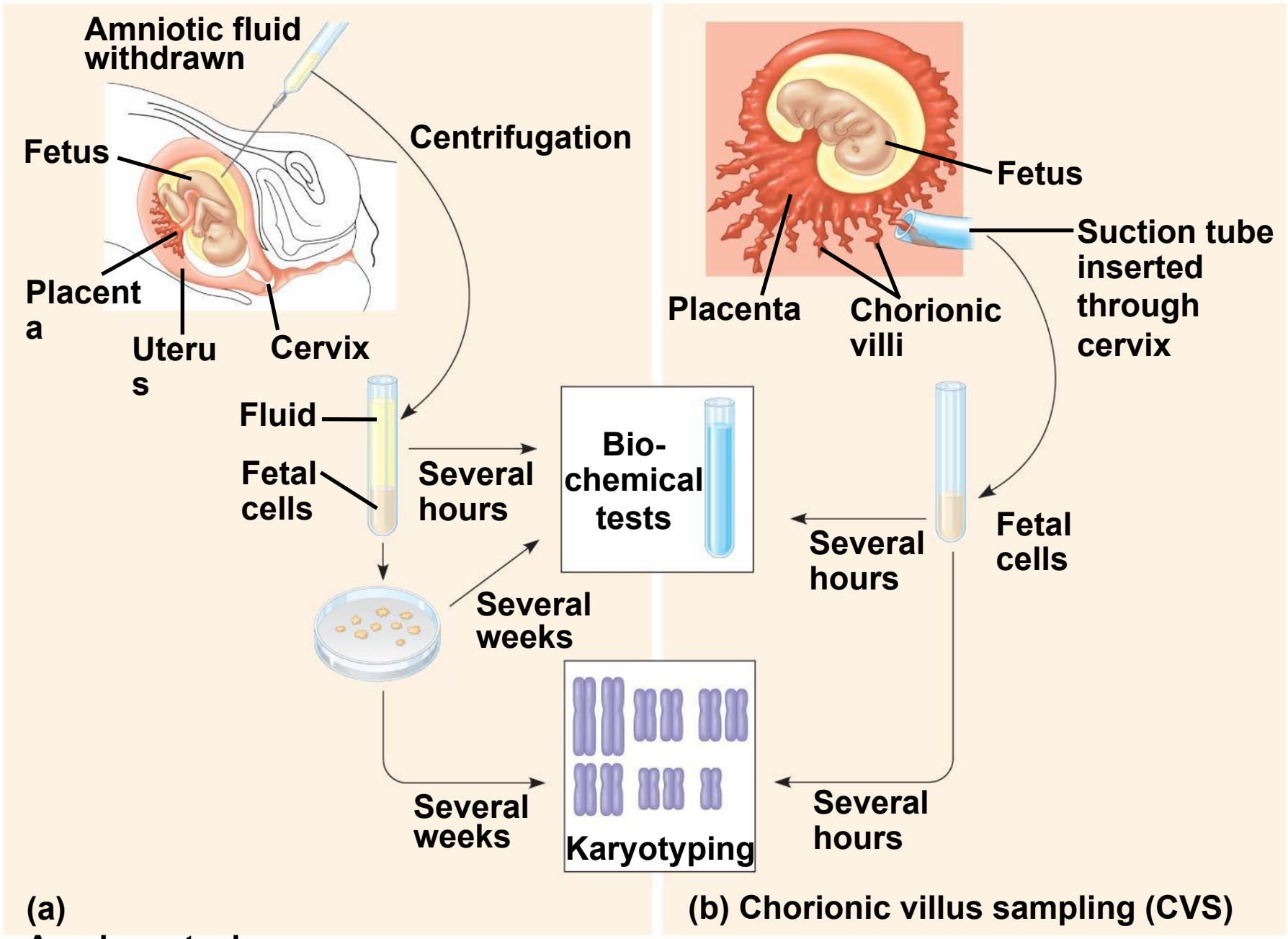

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Fig. 14-18a

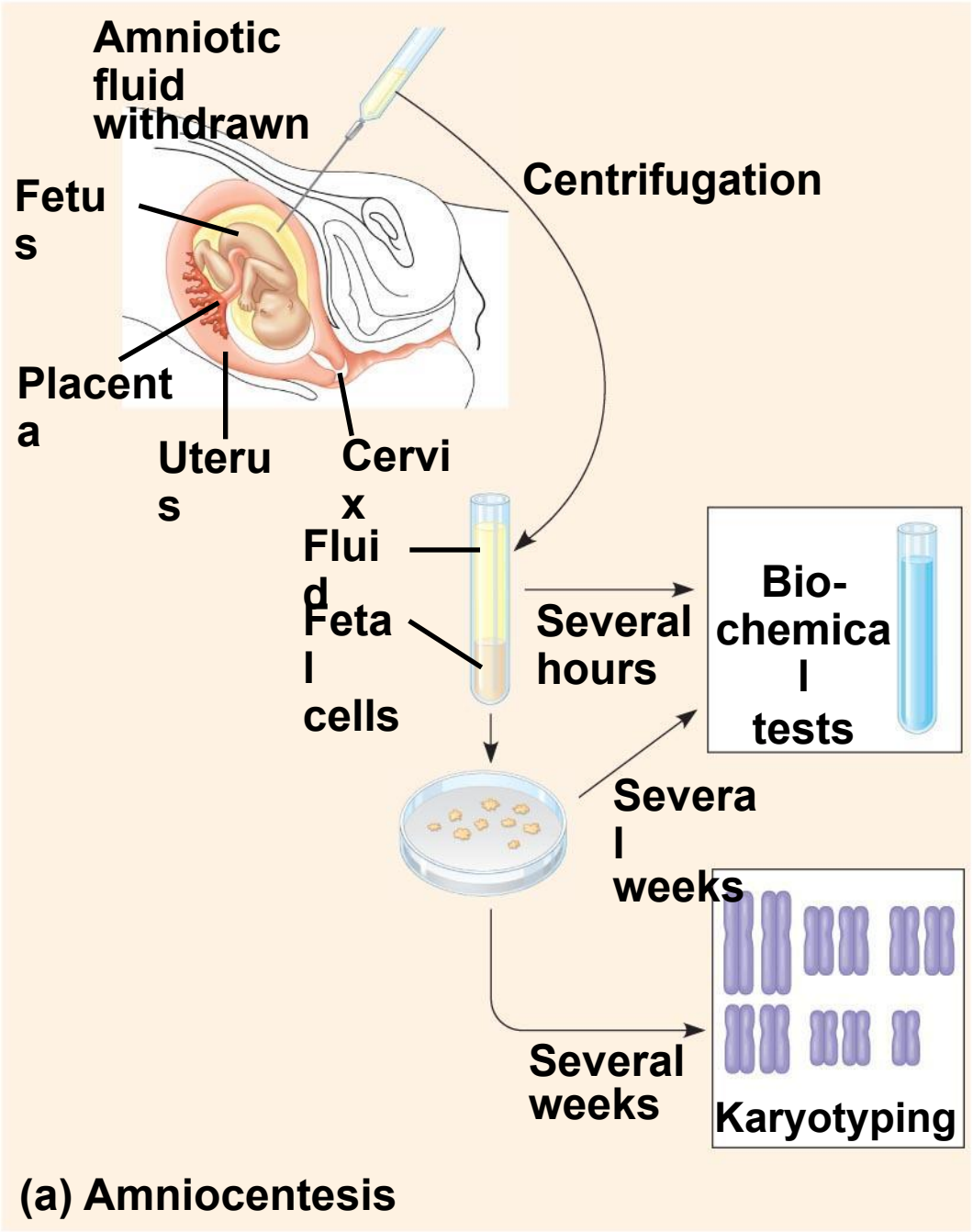

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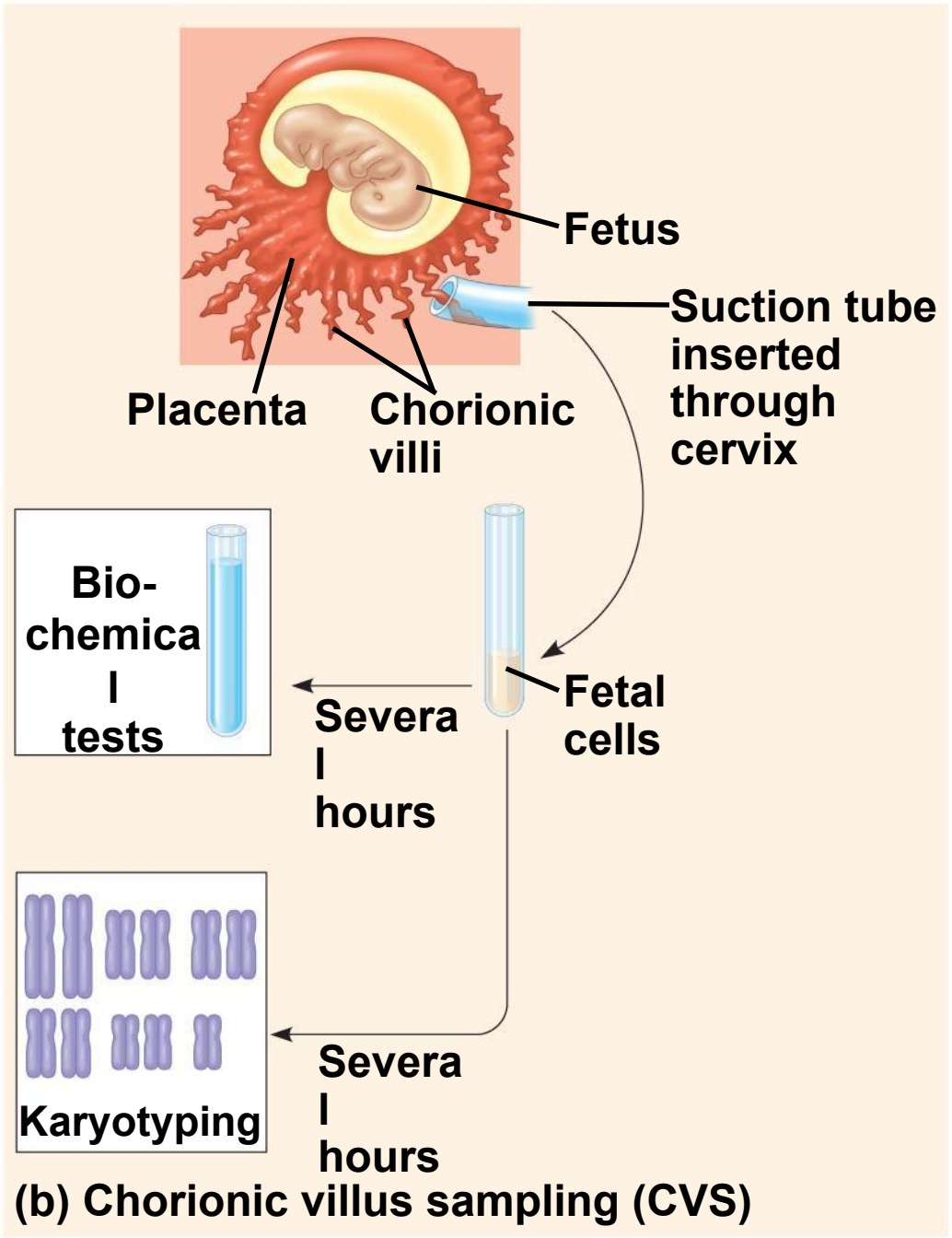
Fig. 14-18b



(b) Chorionic villus sampling (CVS)

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Newborn Screening

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

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Fig. 14-UN2

Degree of dominance	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homozygous dominant	
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	
Codominance	Heterozygotes: Both phenotypes expressed	I^AI^B 
Multiple alleles	In the whole population, some genes have more than two alleles	ABO blood group alleles I^A, I^B, i
Pleiotropy	One gene is able to affect multiple phenotypic characters	Sickle-cell disease

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Fig. 14-UN3

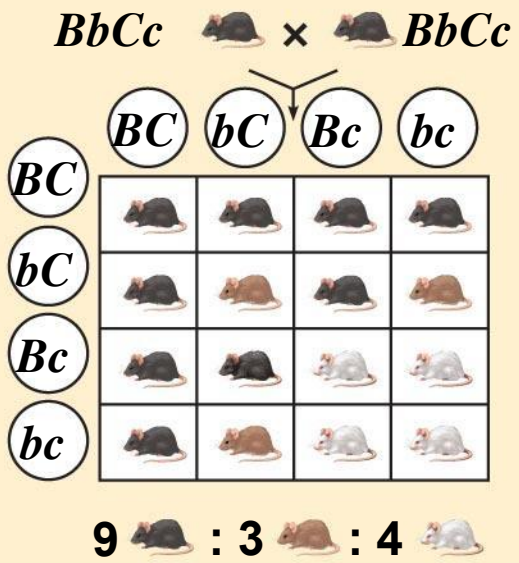
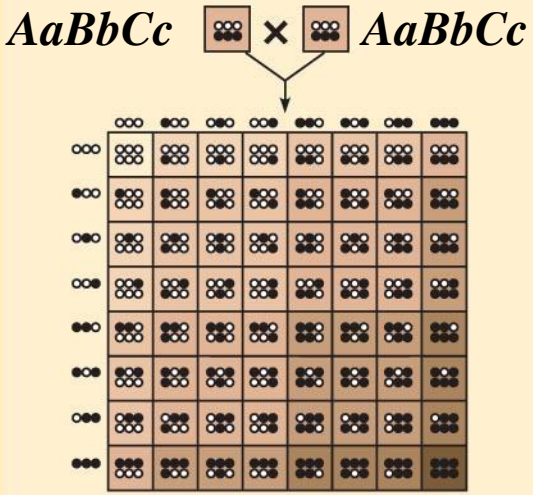
Relationship among genes	Description	Example
Epistasis	One gene affects the expression of another	
Polygenic inheritance	A single phenotypic character is affected by two or more genes	

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Fig. 14-UN4

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Fig. 14-UN5

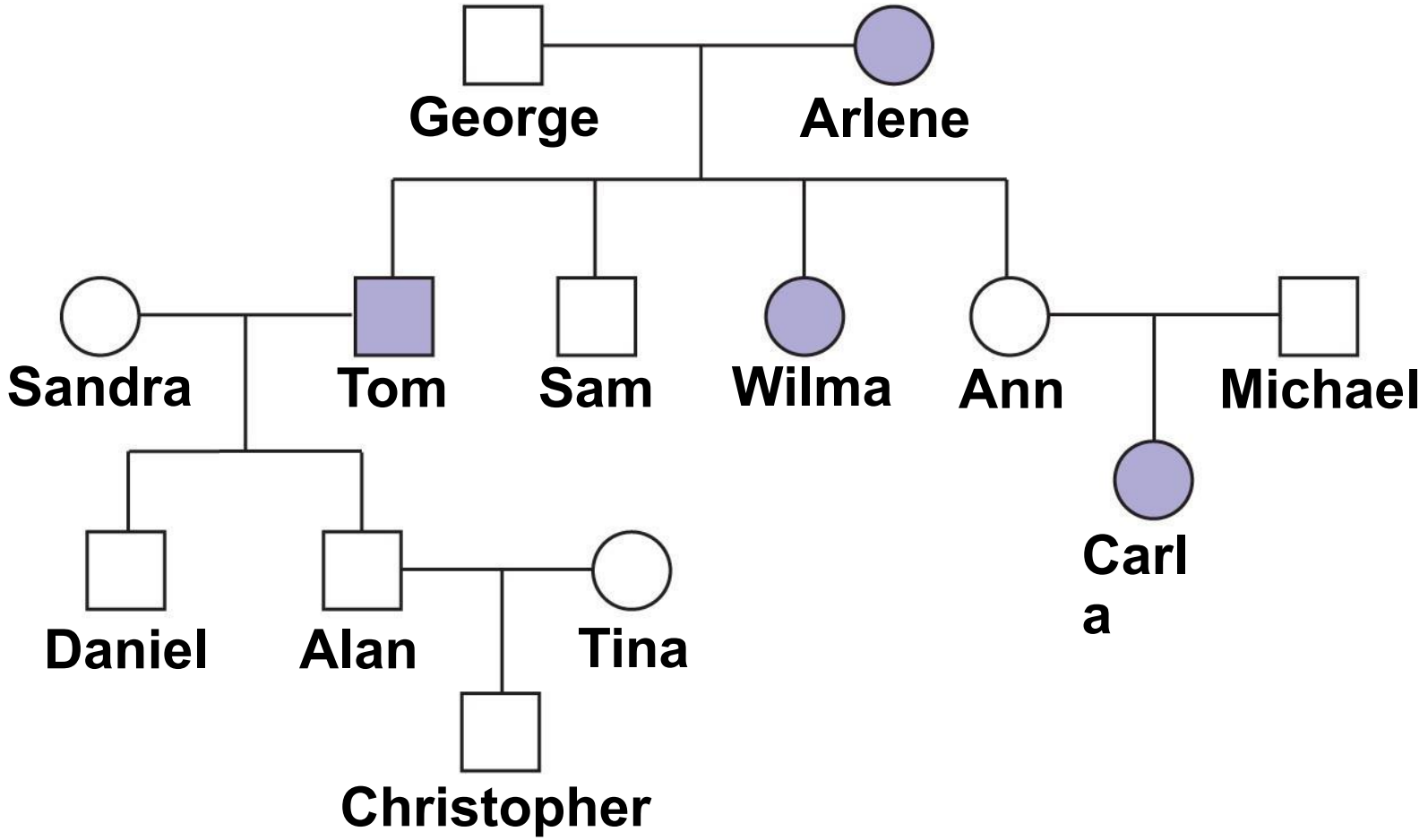

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Fig. 14-UN6

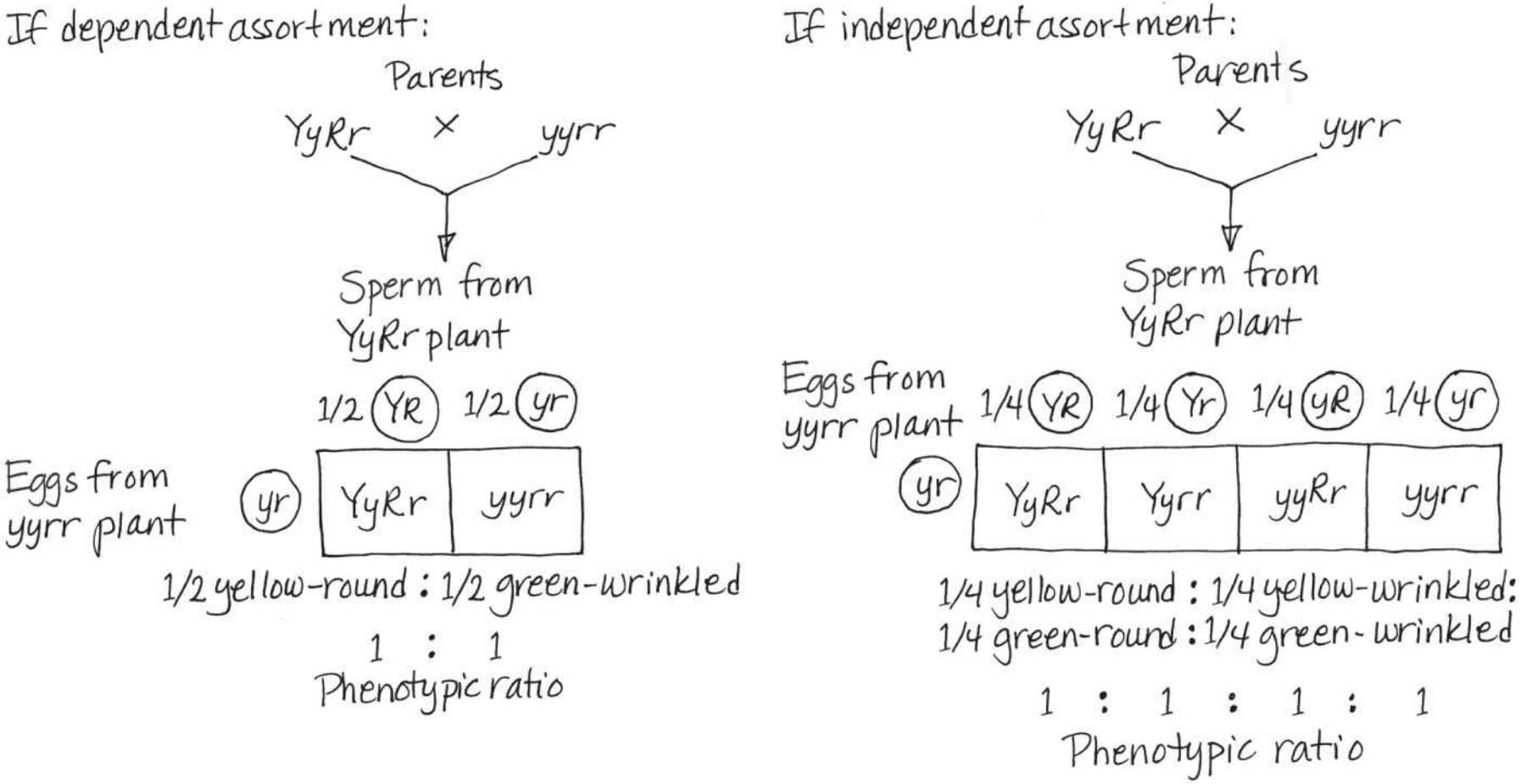

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Fig. 14-UN7

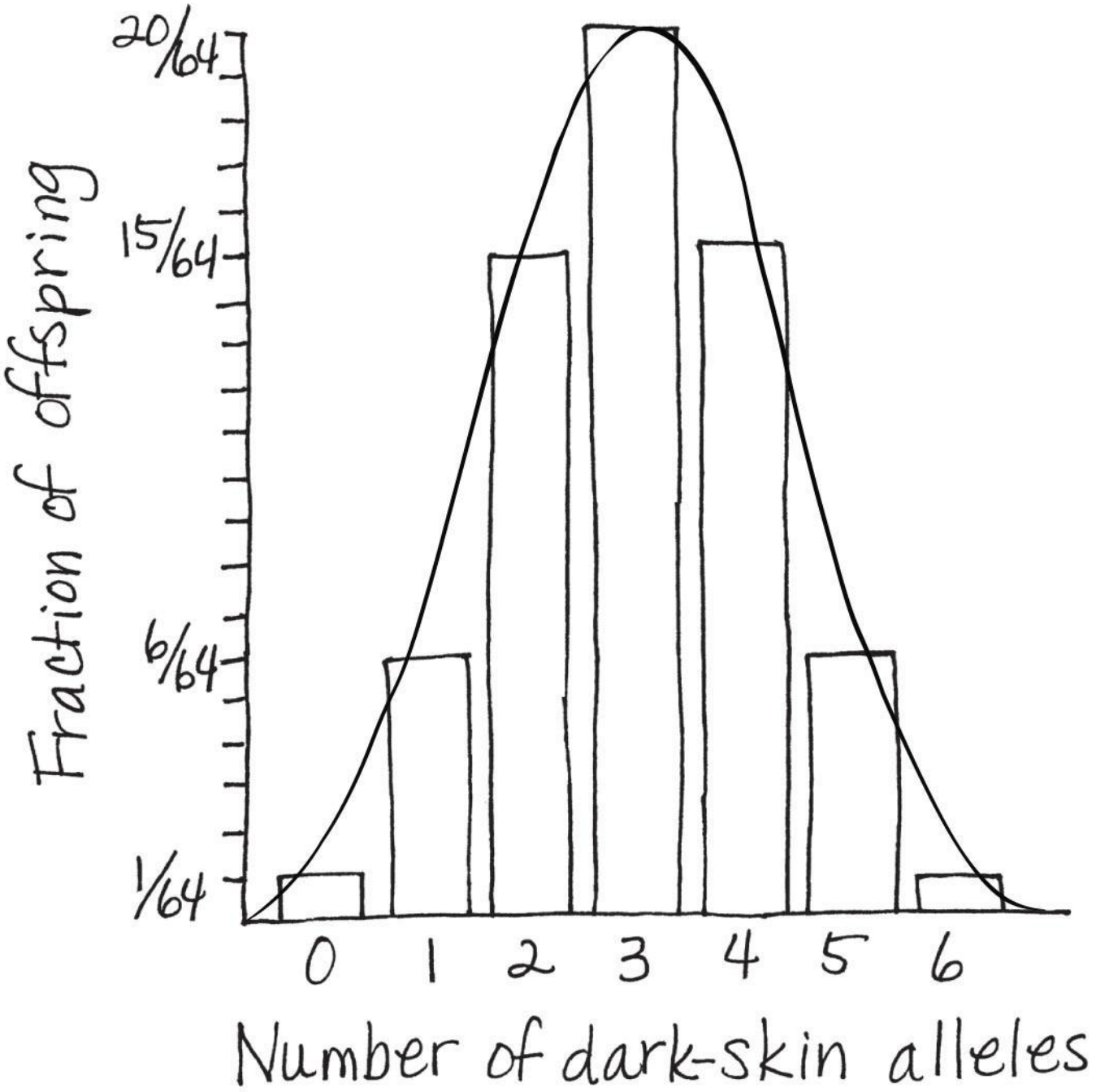

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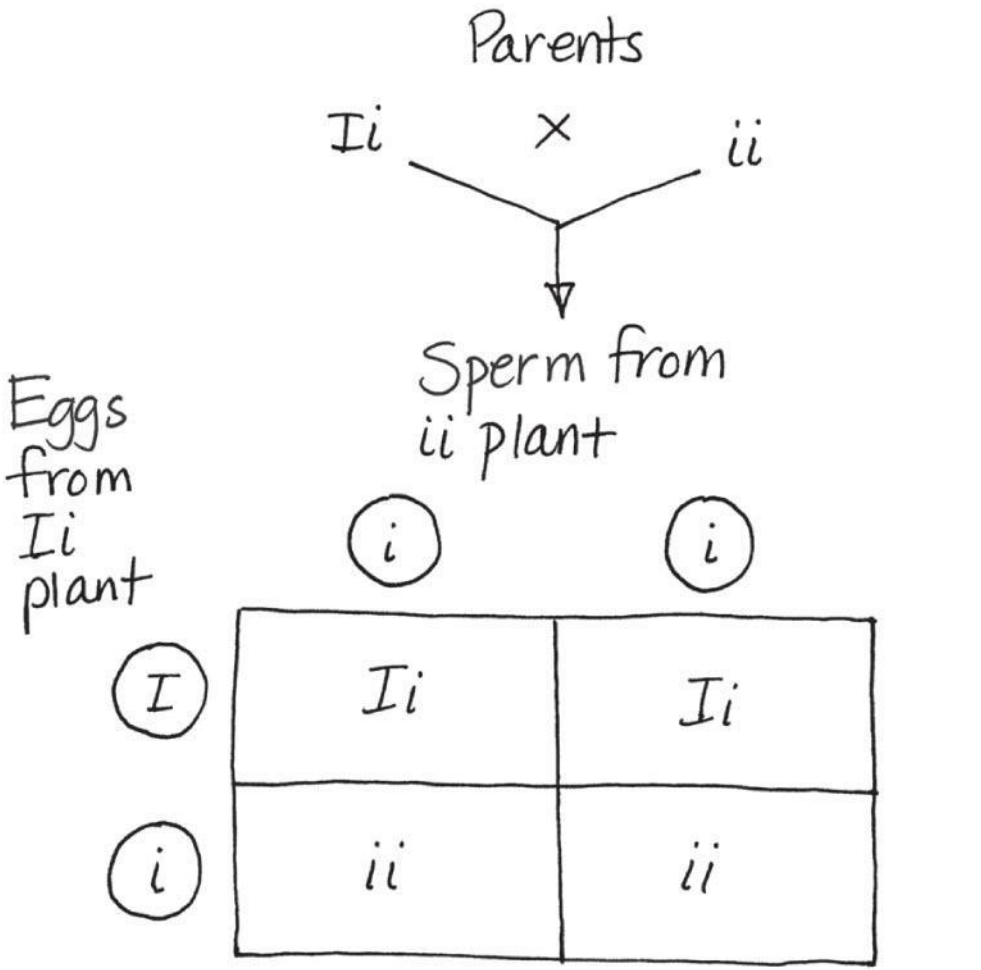
Fig. 14-UN8



Genotypic ratio 1 Ii : 1 ii
(2:2 is equivalent)

Phenotypic ratio 1 inflated : 1 constricted
(2:2 is equivalent)

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Fig. 14-UN9

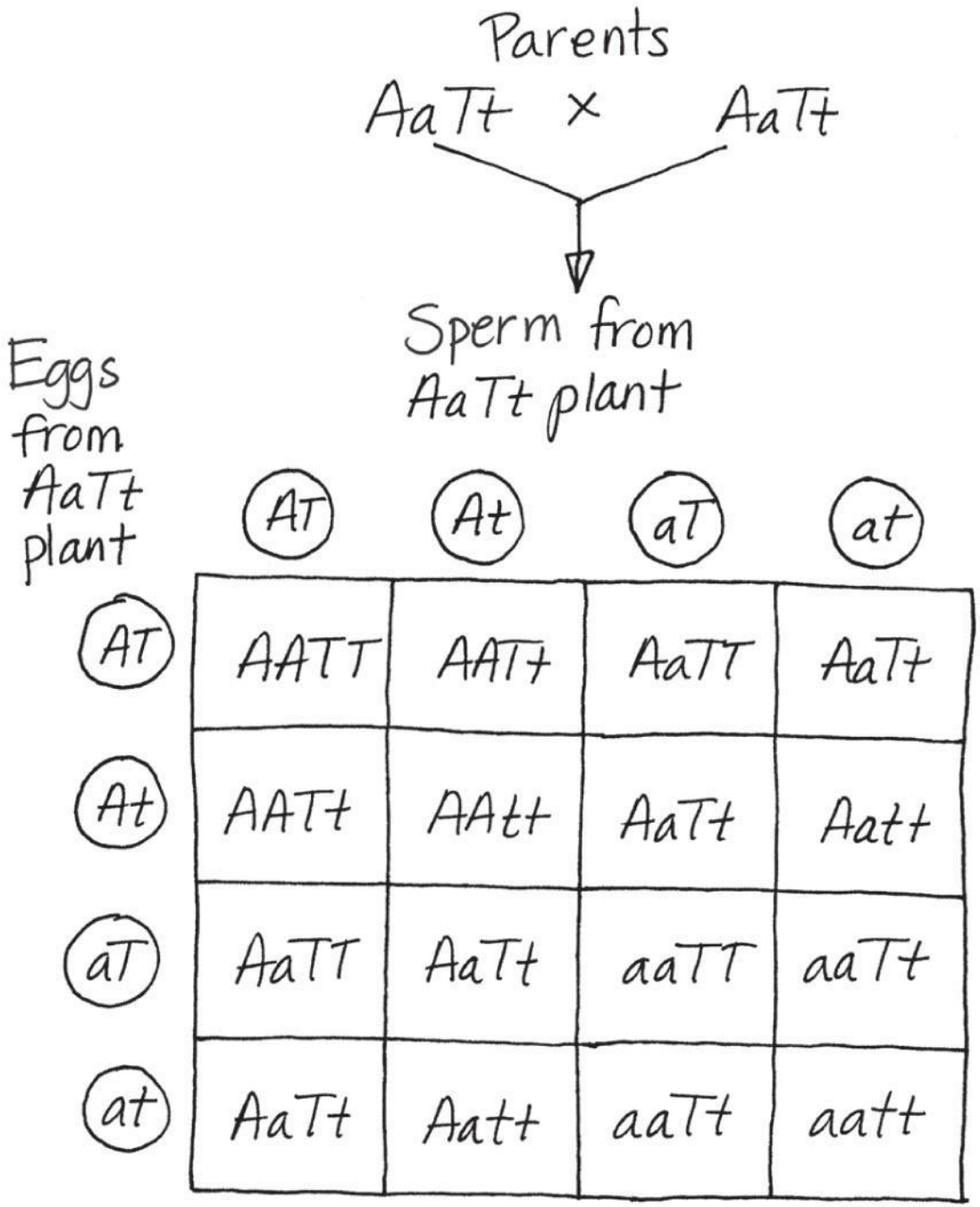

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Fig. 14-UN10

ppyyIi	½ (probability of pp) × ¼ (yy) × ½ (ii)	= 1/16
ppYyii	½ (pp) × ½ (Yy) × ½ (ii)	= 2/16
Ppyyii	½ (Pp) × ¼ (yy) × ½ (ii)	= 1/16
ppYYii	½ (pp) × ¼ (YY) × ½ (ii)	= 1/16
ppyyii	½ (pp) × ¼ (yy) × ½ (ii)	= 1/16
Fraction predicted to have at least two recessive traits		= 6/16 or 3/8

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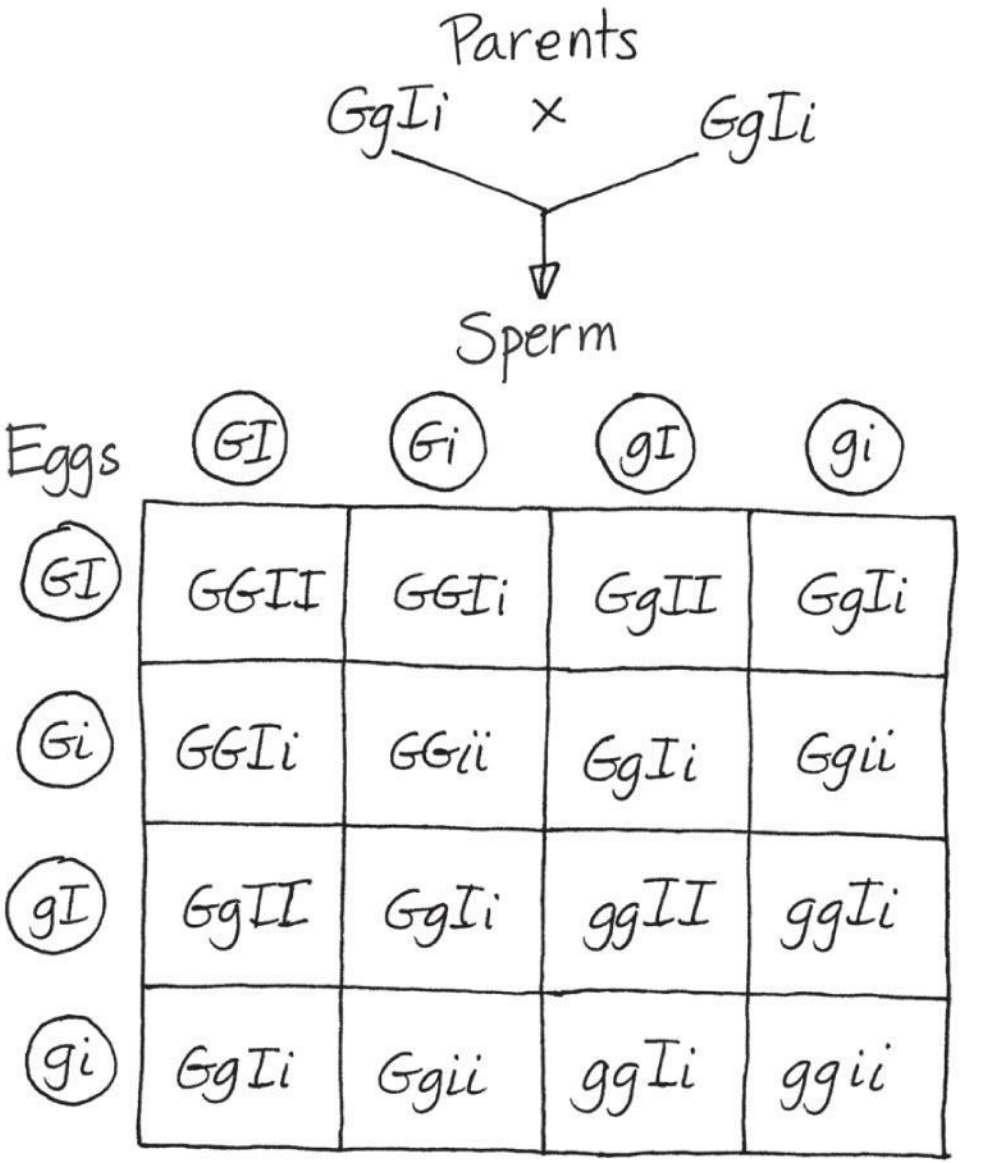
Fig. 14-UN11



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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

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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

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