## 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

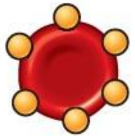
# Multiple Alleles

- 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, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease

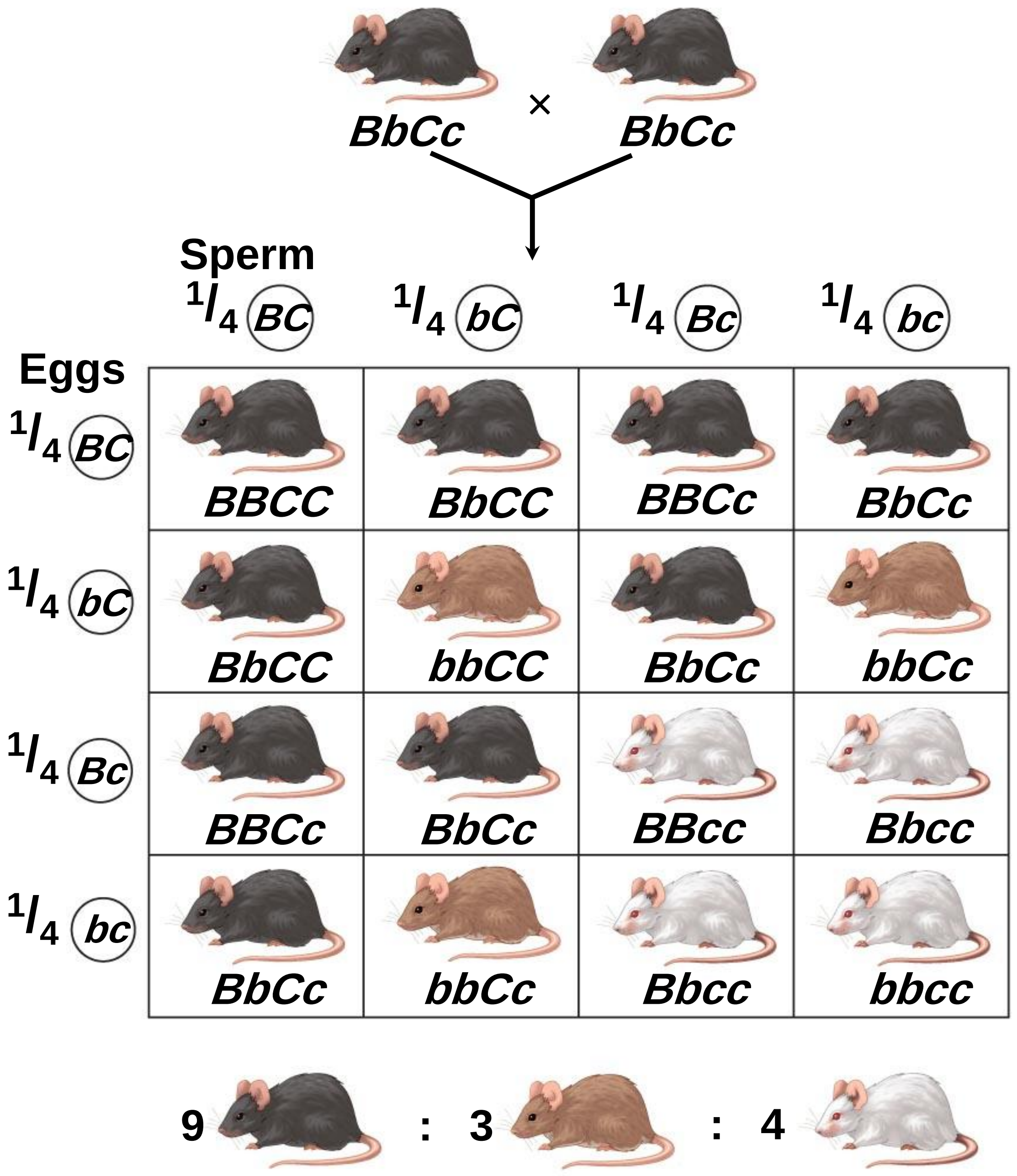
# Extending Mendelian Genetics for Two or More Genes

- 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
- 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

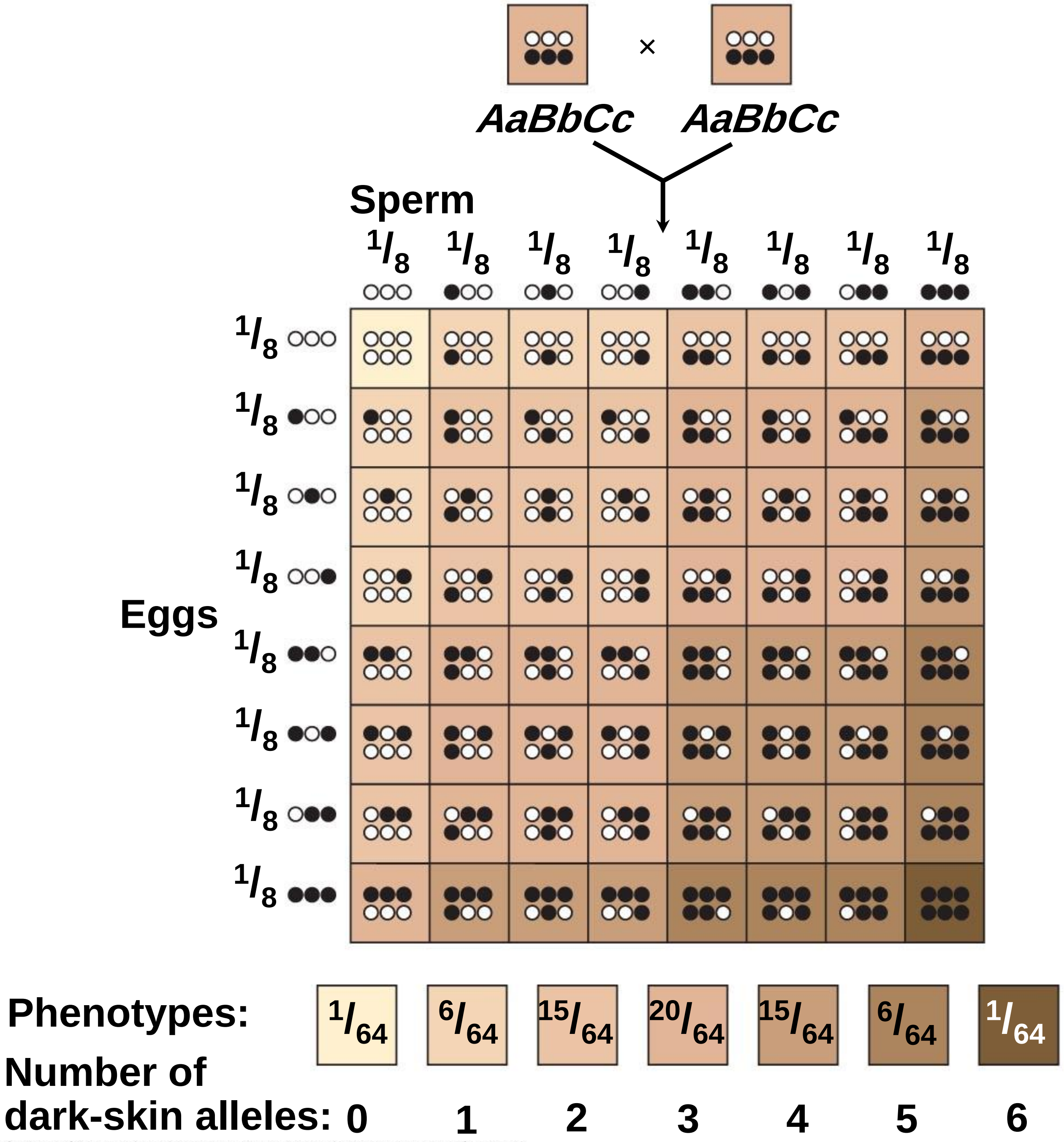
Fig. 14-12


BbCc
×
BbCc
Sperm
1/4 BC
1/4 bC
1/4 Bc
1/4 bc
Eggs
1/4 BC
1/4 bC
1/4 Bc
1/4 bc
BBCC
BbCC
BBCc
BbCc
BbCC
bbCC
BbCc
bbCc
BBCc
BbCc
BBcc
Bbcc
BbCc
bbCc
Bbcc
bbcc
9
:
3
:
4

# *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


AaBbCc
×
AaBbCc
Sperm
1/8
Eggs
1/8
Phenotypes:
1/64
6/64
15/64
20/64
15/64
6/64
1/64
Number of dark-skin alleles:
0
1
2
3
4
5
6

# 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

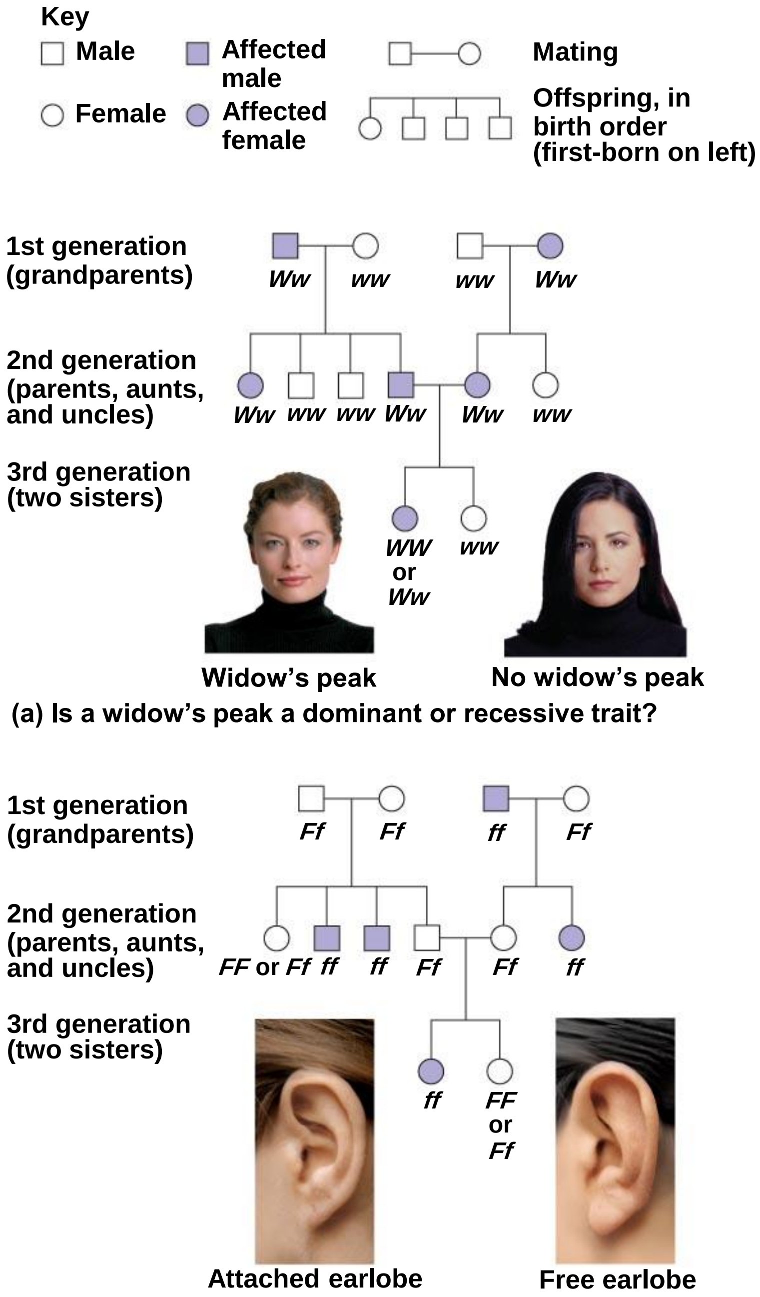
# Concept 14.4: Many human traits follow Mendelian patterns of inheritance

- 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

Fig. 14-15




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

Fig. 14-15a

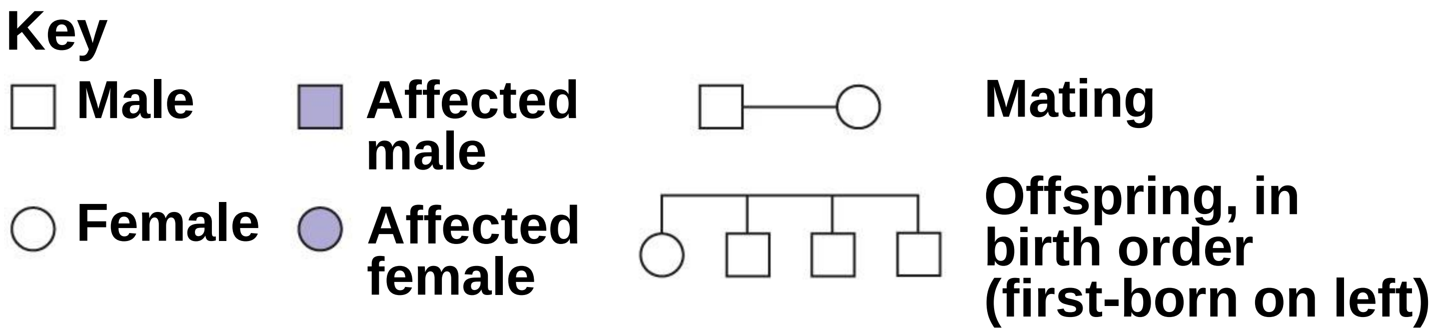
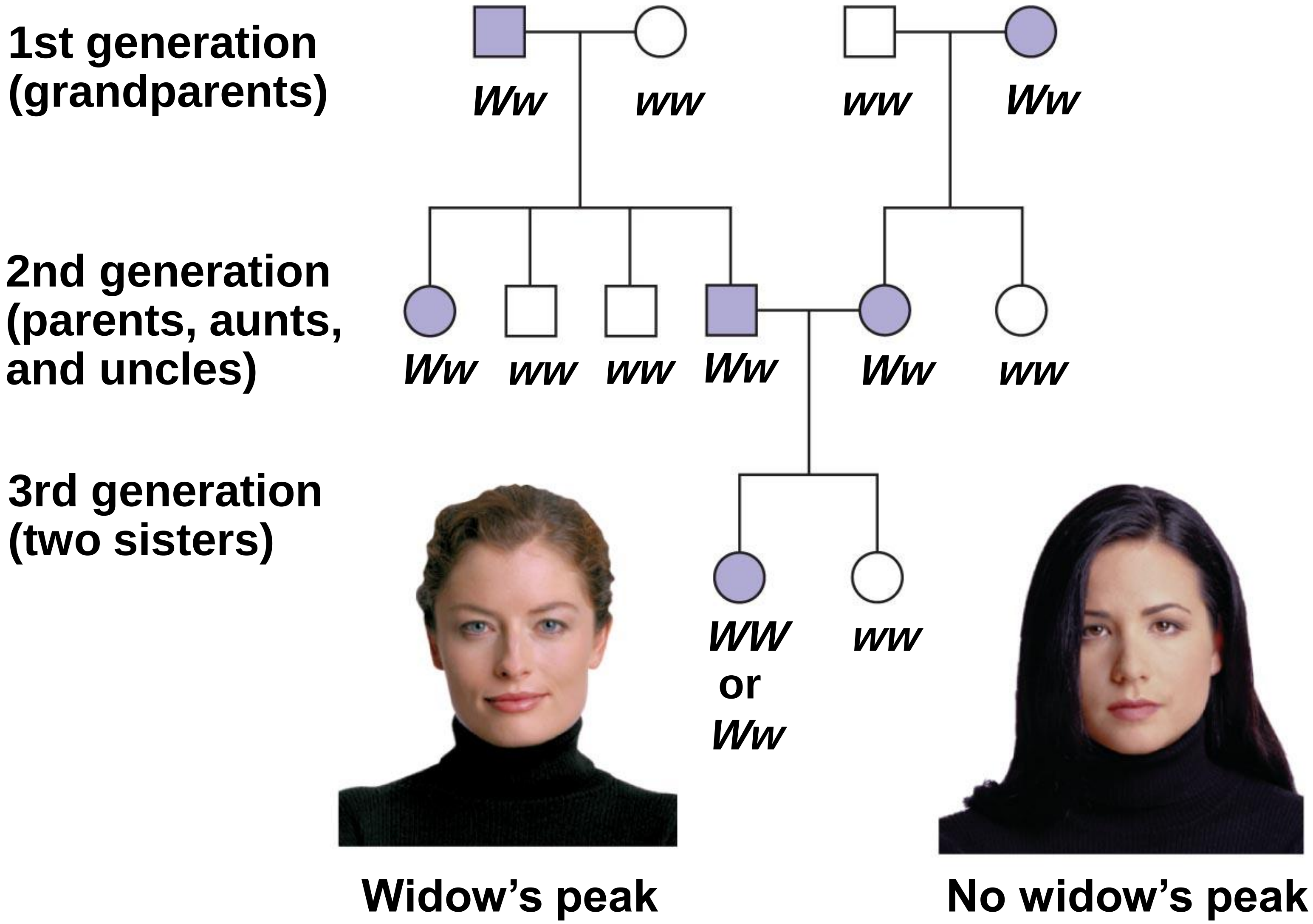
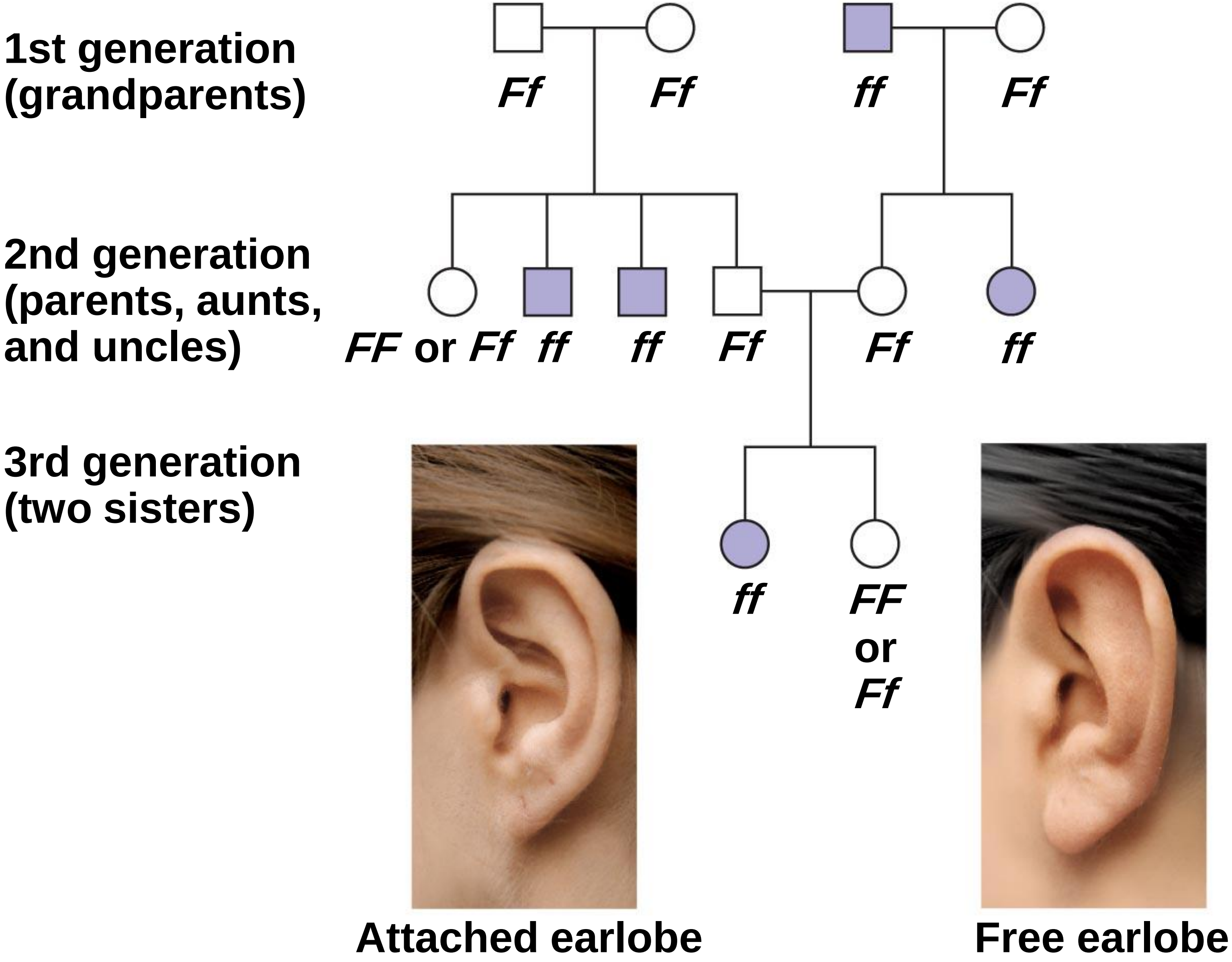

Key
Male
Affected male
Mating
Female
Affected female
Offspring, in birth order (first-born on left)

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

**Parents**

| Normal *Aa* | × | Normal *Aa* |
|---|---|---|

**Sperm**

| Eggs | *A* | *a* |
|---|---|---|
| *A* | *AA* Normal | *Aa* Normal (carrier) |
| *a* | *Aa* Normal (carrier) | *aa* Albino |

- 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* is a form of dwarfism caused by a rare dominant allele

Fig. 14-17

**Parents**

**Dwarf** *Dd* × **Normal** *dd*

**Sperm**

| Eggs | *D* | *d* |
| --- | --- | --- |
| *d* | *Dd* Dwarf | *dd* Normal |
| *d* | *Dd* Dwarf | *dd* Normal |

# *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

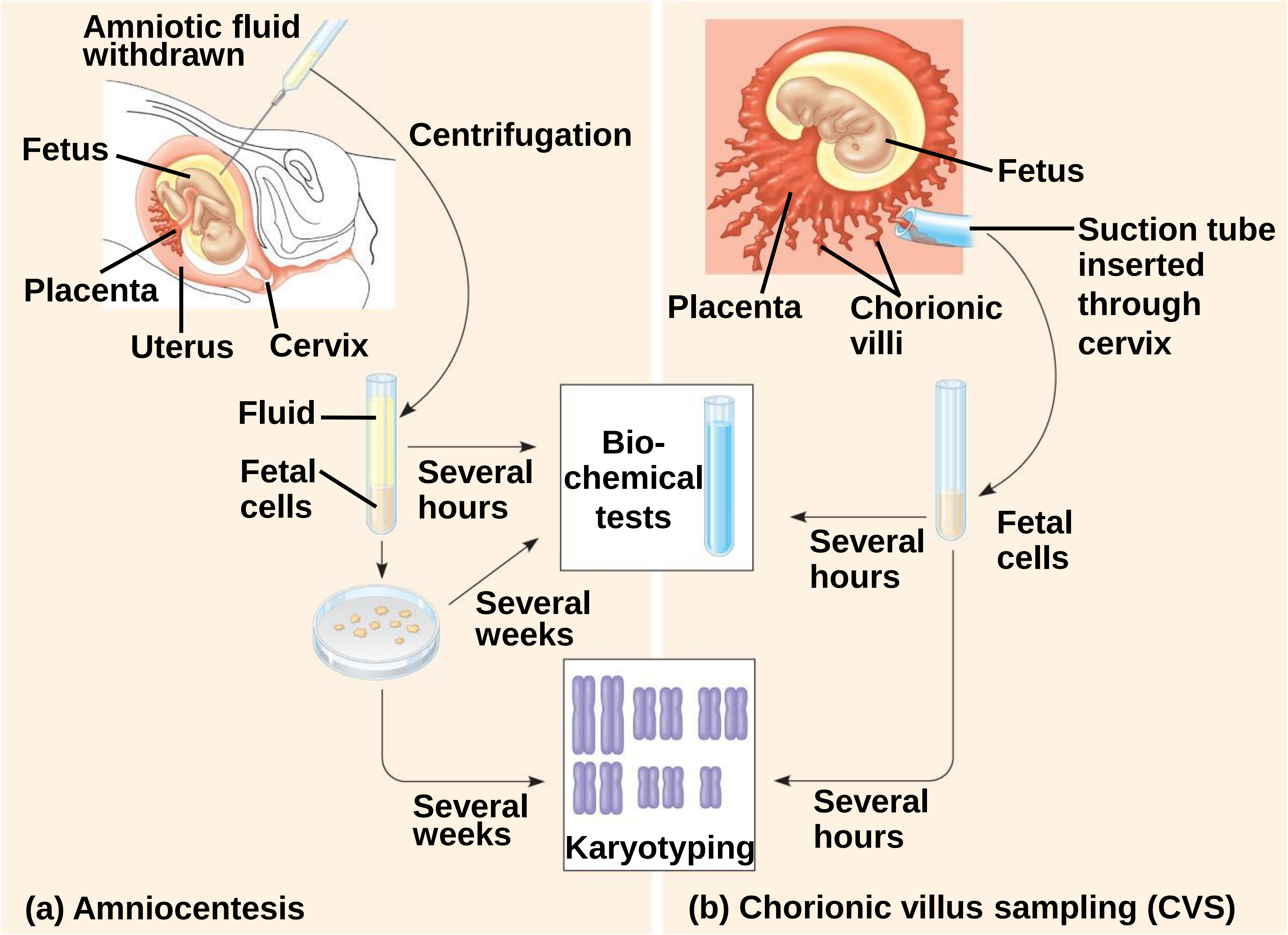

Amniotic fluid withdrawn
Fetus
Placenta
Uterus
Cervix
Centrifugation
Fluid
Fetal cells
Several hours
Bio-chemical tests
Several weeks
Several weeks
Karyotyping
(a) Amniocentesis
Fetus
Suction tube inserted through cervix
Placenta
Chorionic villi
Fetal cells
Several hours
Several hours
(b) Chorionic villus sampling (CVS)

Fig. 14-18a

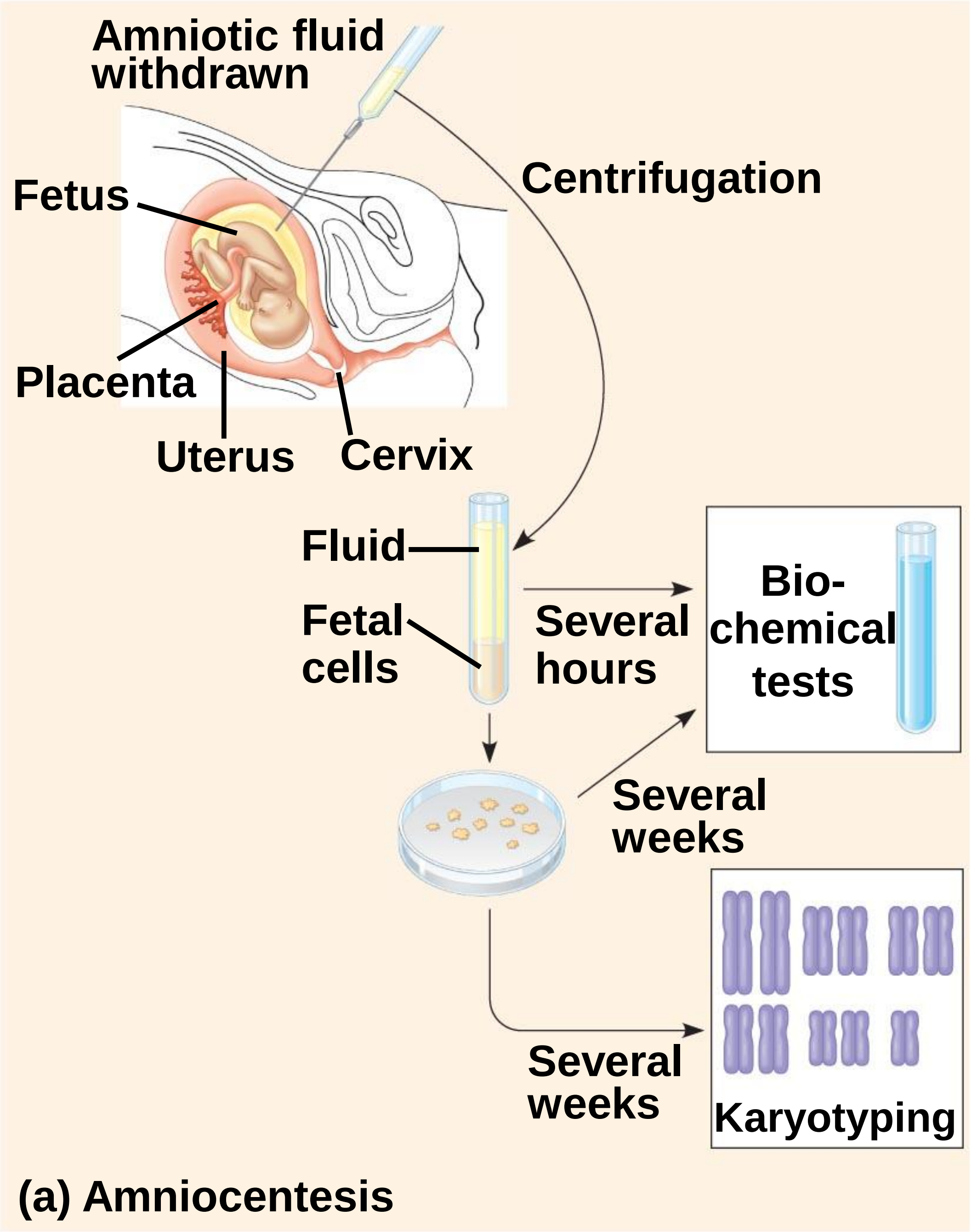
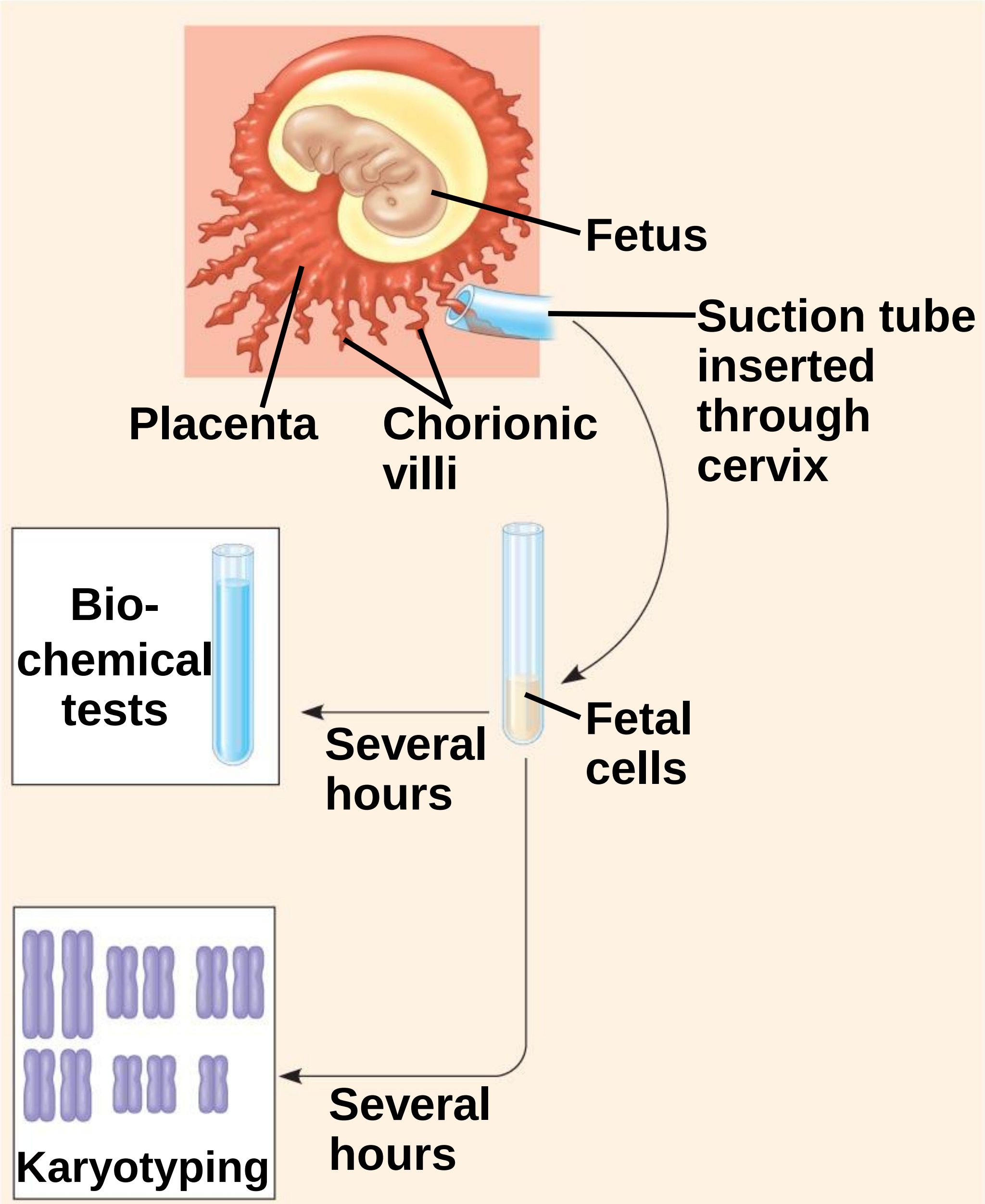

Amniotic fluid withdrawn
Centrifugation
Fetus
Placenta
Uterus
Cervix
Fluid
Fetal cells
Several hours
Bio-chemical tests
Several weeks
Several weeks
Karyotyping
(a) Amniocentesis

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 in the United States

Fig. 14-UN2

| Degree of dominance | Description | Example |
|---|---|---|
| Complete dominance of one allele | Heterozygous phenotype same as that of homo-zygous dominant | $PP$  $Pp$  |
| Incomplete dominance of either allele | Heterozygous phenotype intermediate between the two homozygous phenotypes |  $C^RC^R$ $C^RC^W$ $C^WC^W$ |
| 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 |

Fig. 14-UN3

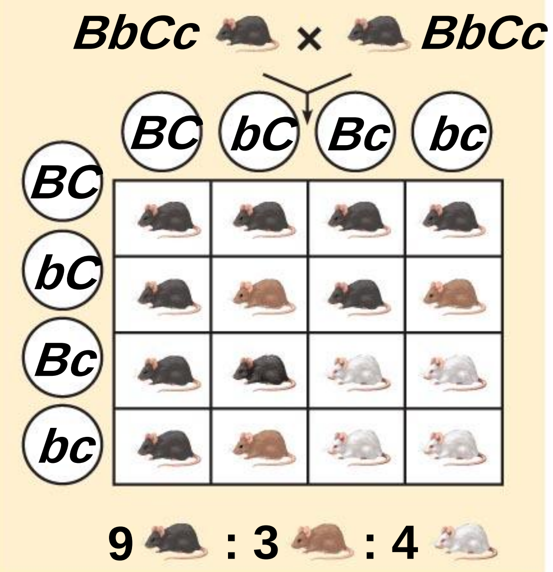
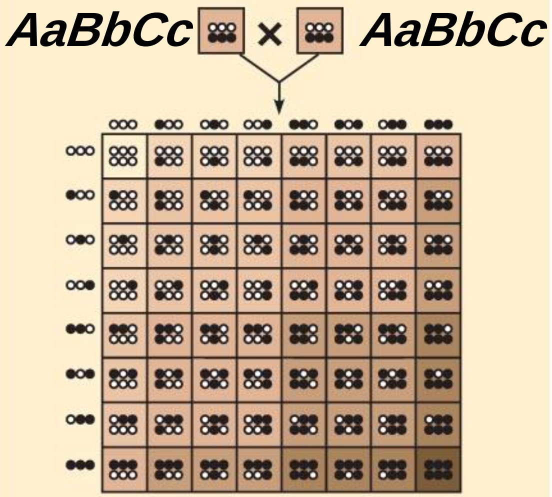
| Relationship among genes | Description | Example |
|---|---|---|
| Epistasis | One gene affects the expression of another |   |
| Polygenic inheritance | A single phenotypic character is affected by two or more genes |   |

Fig. 14-UN4

Fig. 14-UN5

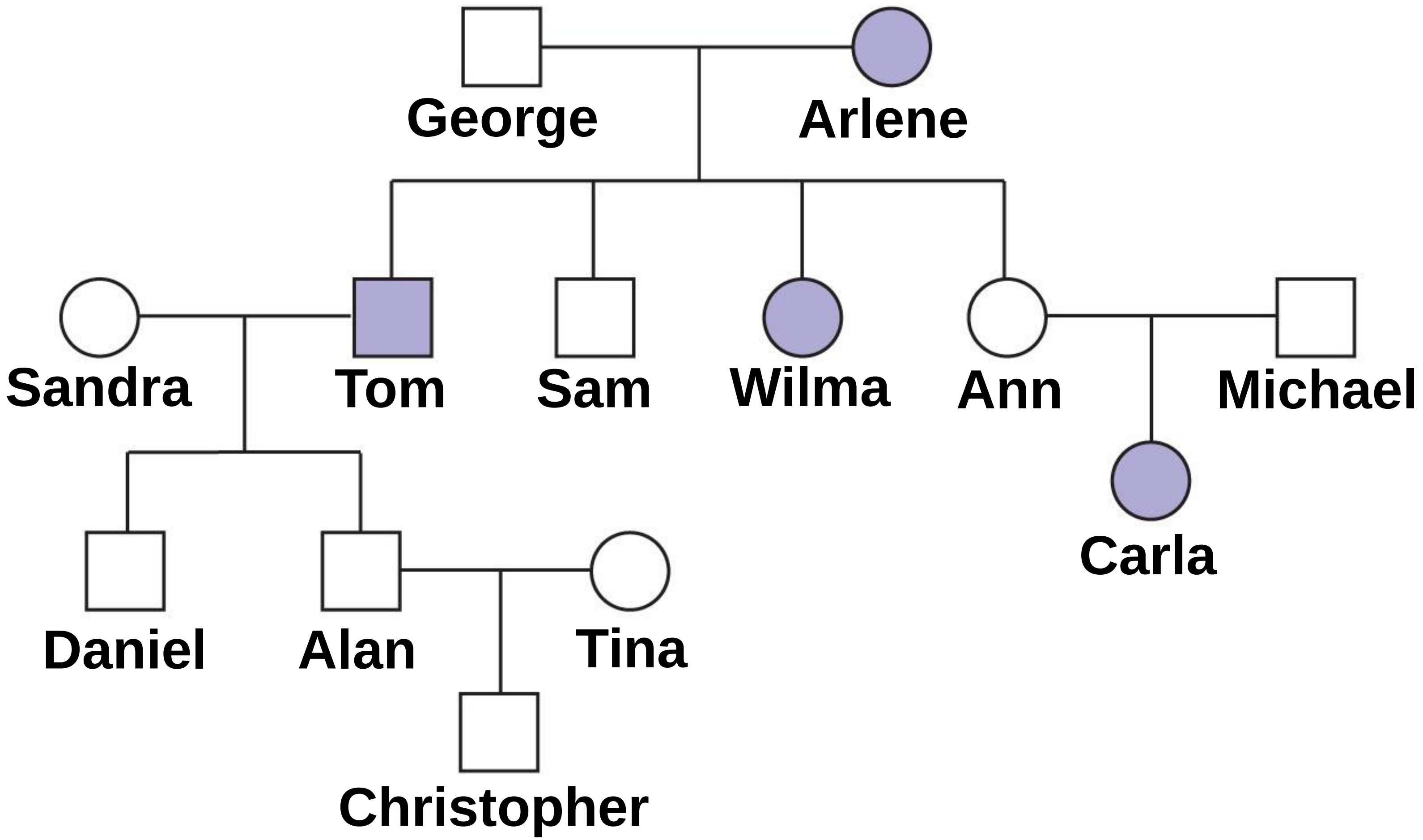
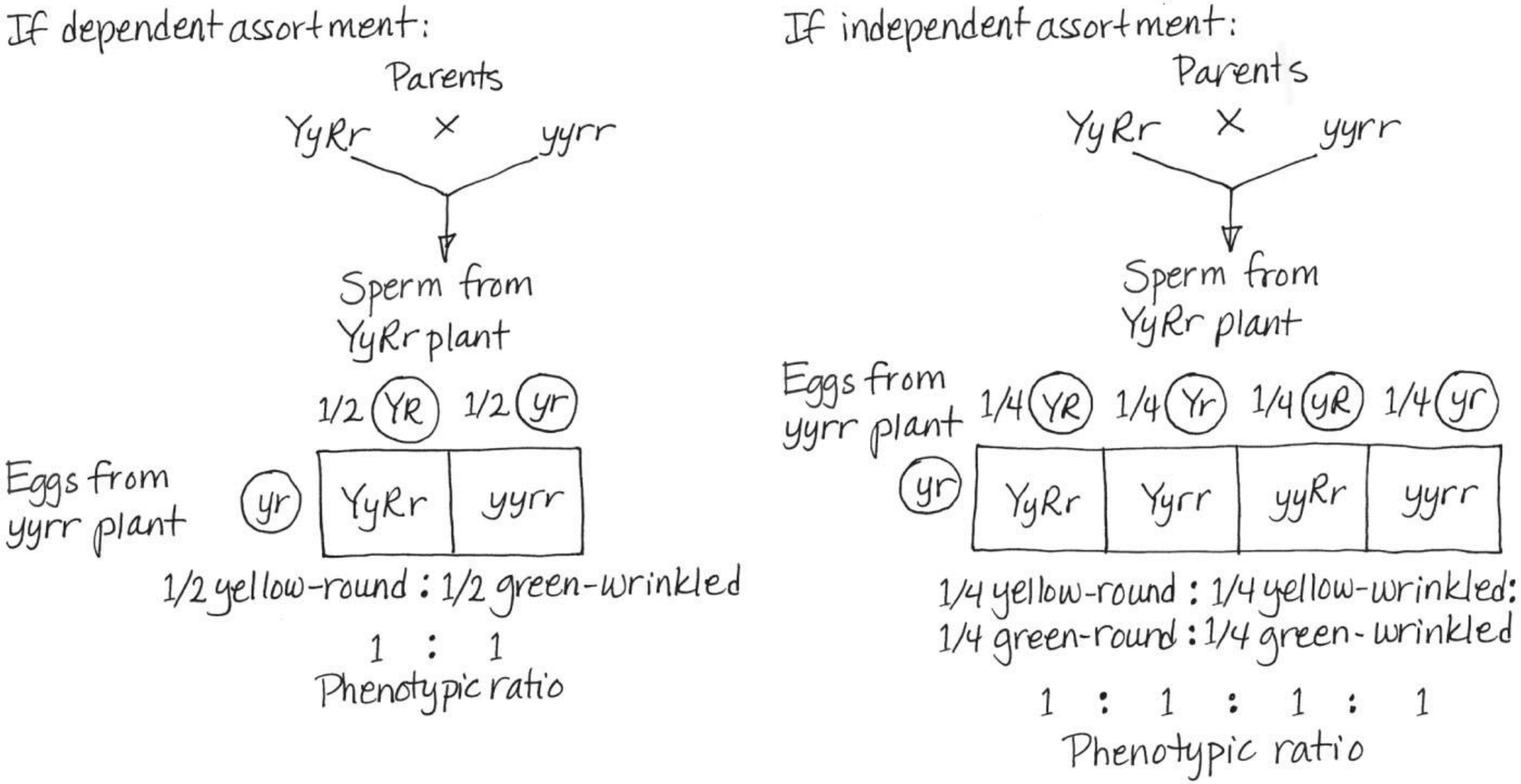
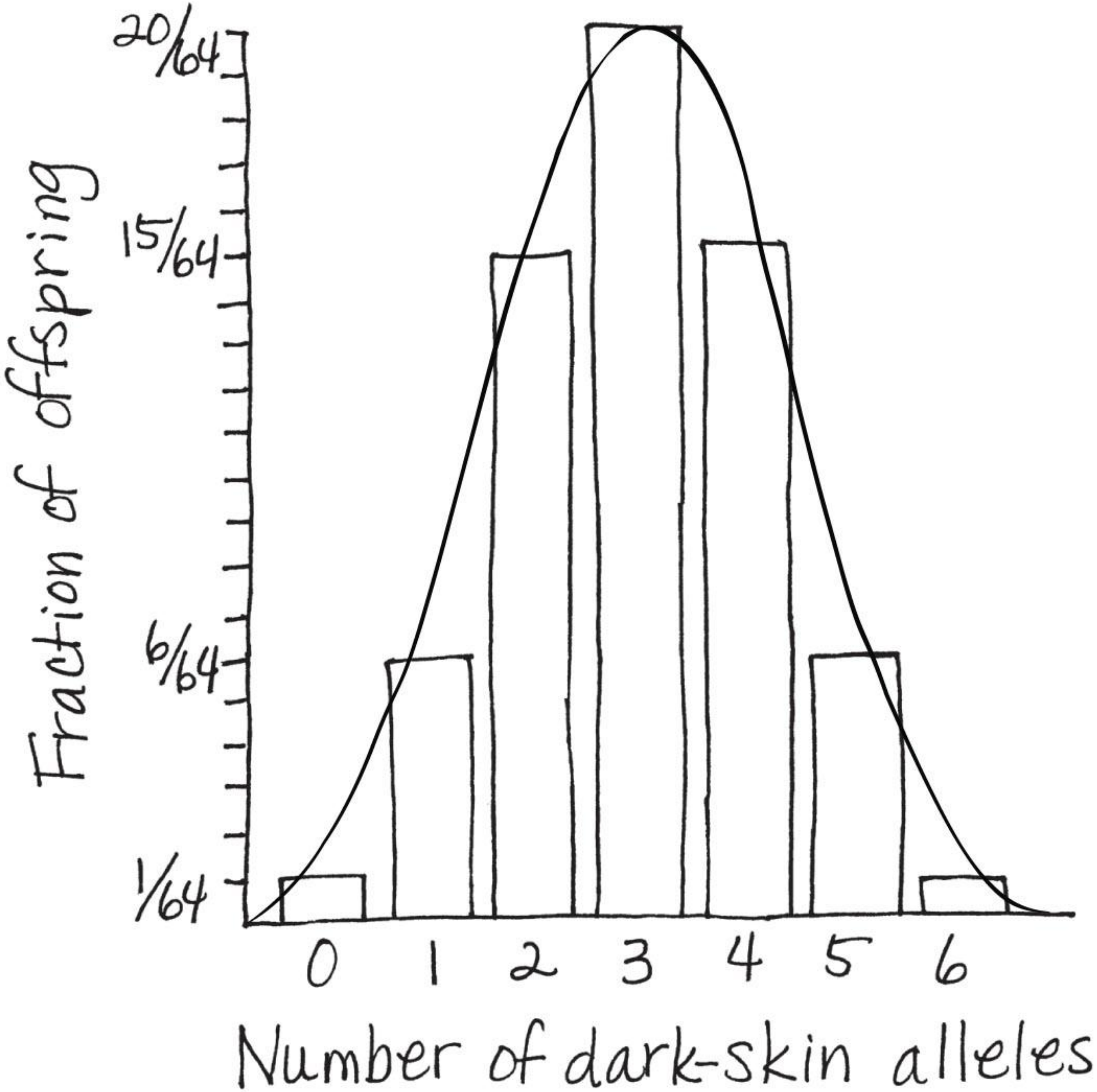

George
Arlene
Sandra
Tom
Sam
Wilma
Ann
Michael
Carla
Daniel
Alan
Tina
Christopher

Fig. 14-UN6



If dependent assortment:
Parents
YyRr
×
yyrr
Sperm from
YyRr plant
1/2
YR
1/2
yr
Eggs from
yyrr plant
yr
YyRr
yyrr
1/2 yellow-round : 1/2 green-wrinkled
1 : 1
Phenotypic ratio
If independent assortment:
Parents
YyRr
×
yyrr
Sperm from
YyRr plant
Eggs from
yyrr plant
1/4
YR
1/4
Yr
1/4
yR
1/4
yr
yr
YyRr
Yyrr
yyRr
yyrr
1/4 yellow-round : 1/4 yellow-wrinkled:
1/4 green-round : 1/4 green-wrinkled
1 : 1 : 1 : 1
Phenotypic ratio

Fig. 14-UN7



20/64
15/64
6/64
1/64
Fraction of offspring
0
1
2
3
4
5
6
Number of dark-skin alleles

Fig. 14-UN8

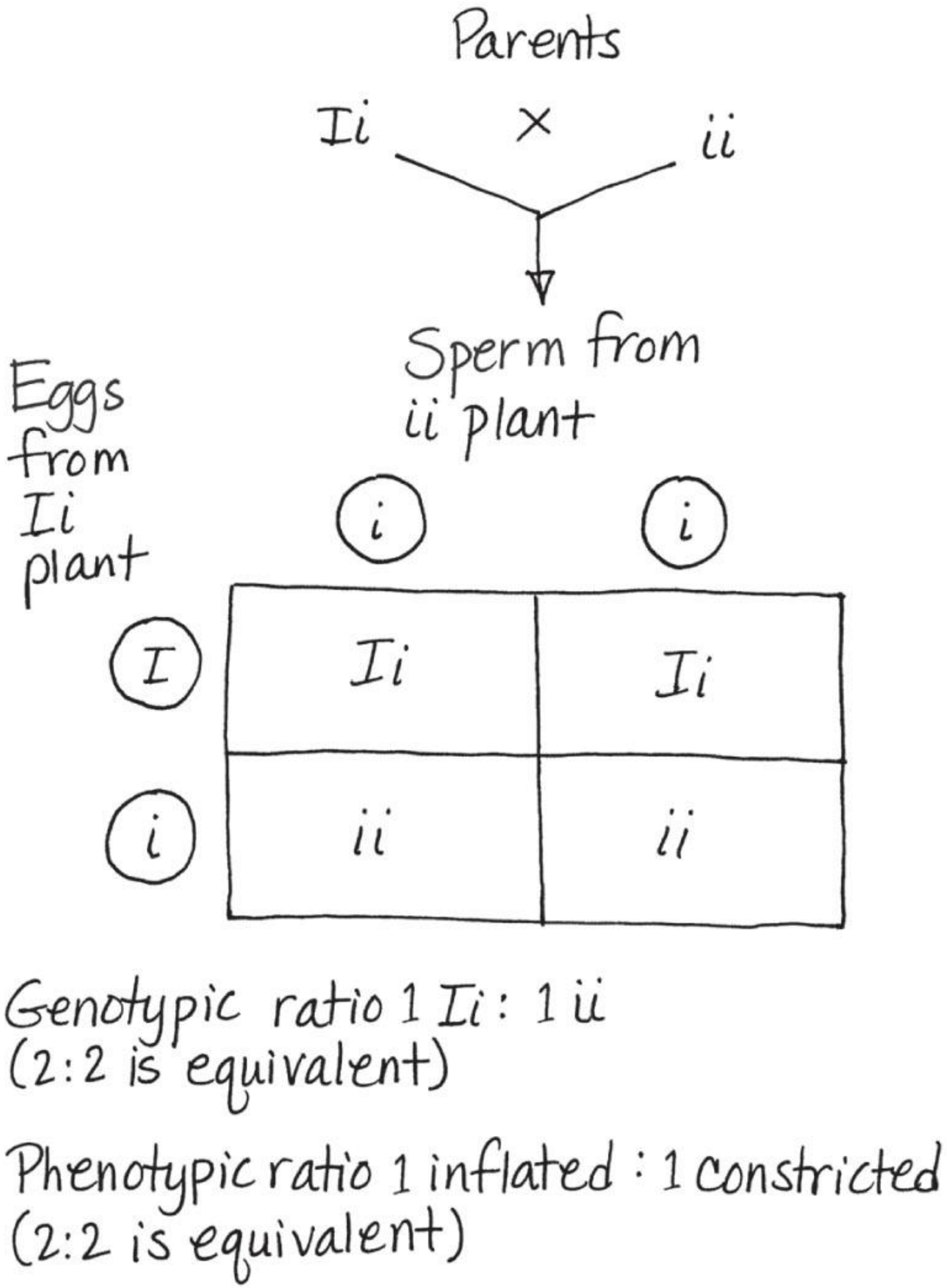
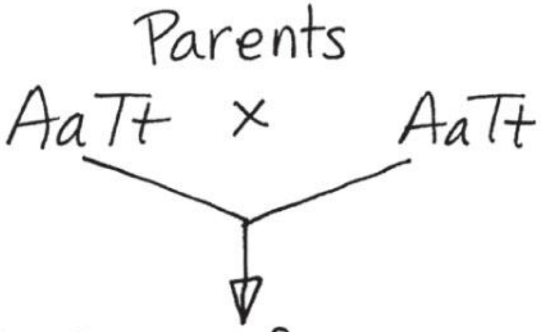

Parents
Ii × ii
Sperm from ii plant
Eggs from Ii plant
i
i
I
i
Ii
Ii
ii
ii
Genotypic ratio 1 Ii : 1 ii
(2:2 is equivalent)
Phenotypic ratio 1 inflated : 1 constricted
(2:2 is equivalent)

Fig. 14-UN9




Sperm from AaTt plant

Eggs from AaTt plant

| | AT | At | aT | at |
|---|---|---|---|---|
| AT | AATT | AATt | AaTT | AaTt |
| At | AATt | AAtt | AaTt | Aatt |
| aT | AaTT | AaTt | aaTT | aaTt |
| at | AaTt | Aatt | aaTt | aatt |

Fig. 14-UN10

| | | |
|---|---|---|
| ppyyIi | $\frac{1}{2}$ (probability of pp) × $\frac{1}{4}$ (yy) × $\frac{1}{2}$ (ii) | $= \frac{1}{16}$ |
| ppYyii | $\frac{1}{2}$ (pp) × $\frac{1}{2}$ (Yy) × $\frac{1}{2}$ (ii) | $= \frac{2}{16}$ |
| Ppyyii | $\frac{1}{2}$ (Pp) × $\frac{1}{4}$ (yy) × $\frac{1}{2}$ (ii) | $= \frac{1}{16}$ |
| ppYYii | $\frac{1}{2}$ (pp) × $\frac{1}{4}$ (YY) × $\frac{1}{2}$ (ii) | $= \frac{1}{16}$ |
| ppyyii | $\frac{1}{2}$ (pp) × $\frac{1}{4}$ (yy) × $\frac{1}{2}$ (ii) | $= \frac{1}{16}$ |

Fraction predicted to have at least two recessive traits $= \frac{6}{16}$ or $\frac{3}{8}$

Fig. 14-UN11

Parents

GgIi × GgIi

Sperm

| Eggs | GI | Gi | gI | gi |
|---|---|---|---|---|
| GI | GGII | GGIi | GgII | GgIi |
| Gi | GGIi | GGii | GgIi | Ggii |
| gI | GgII | GgIi | ggII | ggIi |
| gi | GgIi | Ggii | ggIi | ggii |

9 green-inflated : 3 green-constricted :
3 yellow-inflated : 1 yellow-constricted

## 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