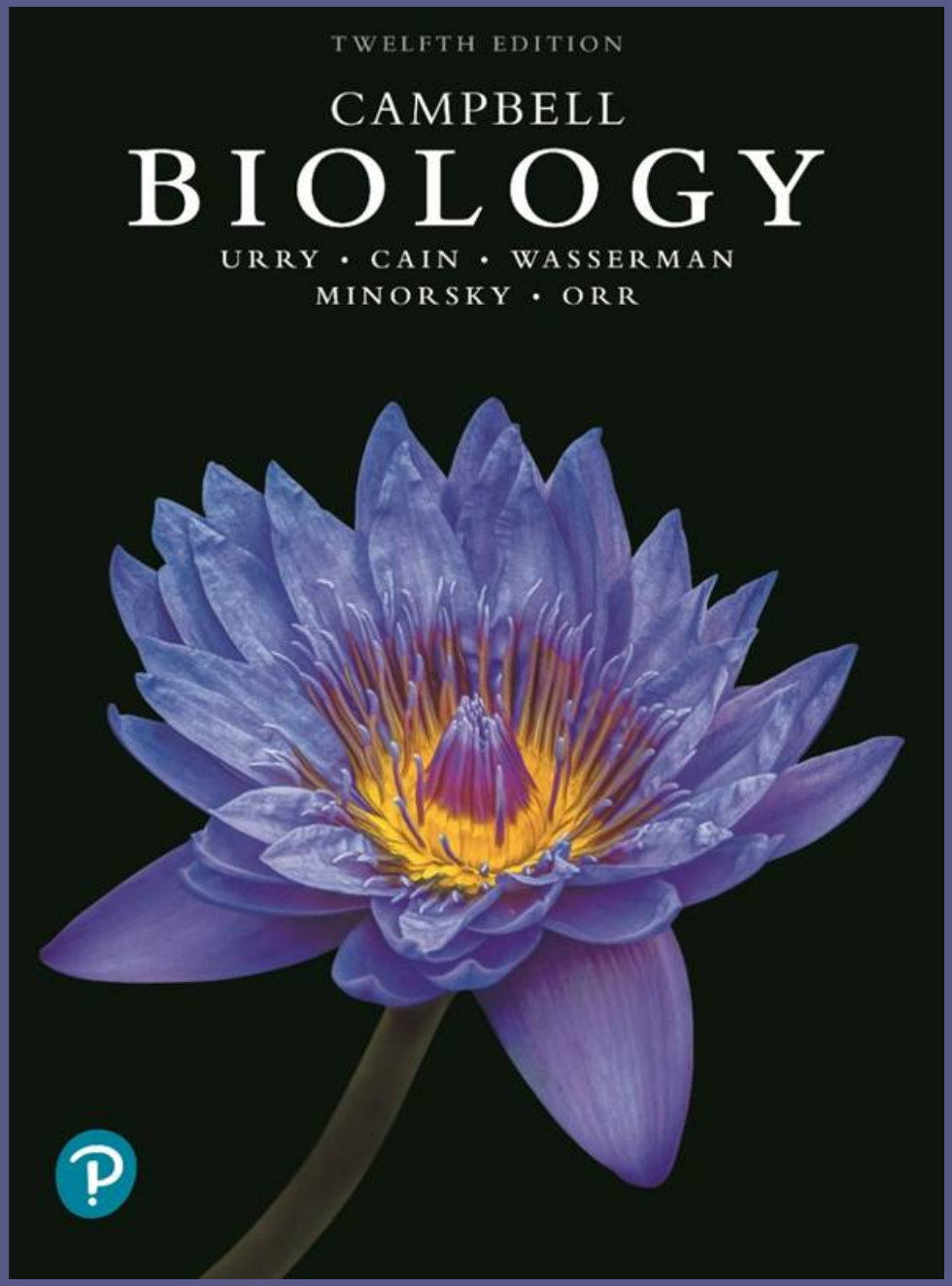


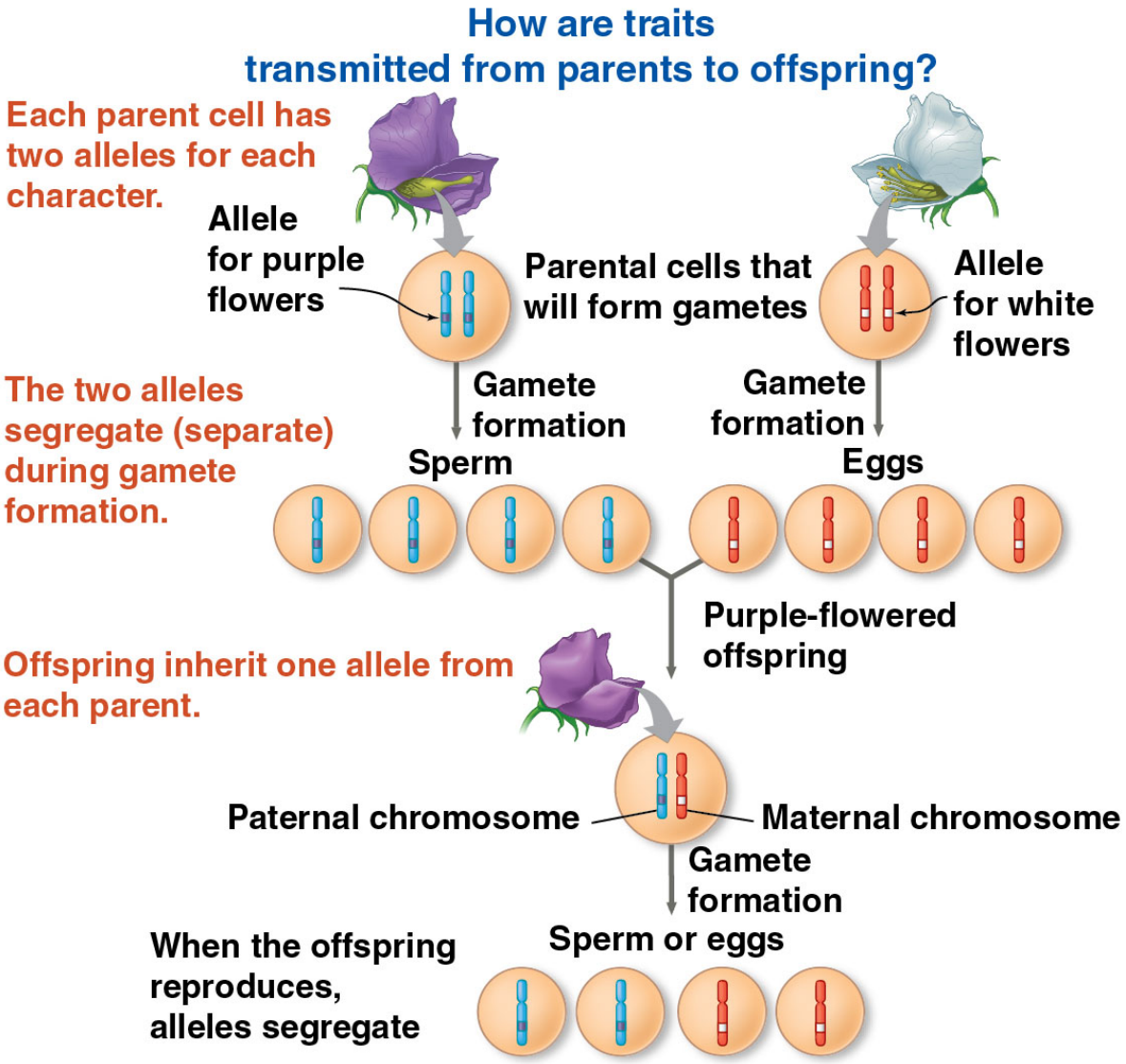
Chapter 14

Mendel and the Gene Idea

Lecture Presentations by
Nicole Tunbridge and
Kathleen Fitzpatrick

Figure 14.1

Figure 14.1a


How are traits transmitted from parents to offspring?
Each parent cell has two alleles for each character.
Allele for purple flowers
Parental cells that will form gametes
Allele for white flowers
The two alleles segregate (separate) during gamete formation.
Gamete formation
Gamete formation
Sperm
Eggs
Purple-flowered offspring
Offspring inherit one allele from each parent.
Paternal chromosome
Maternal chromosome
Gamete formation
When the offspring reproduces, alleles segregate
Sperm or eggs

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

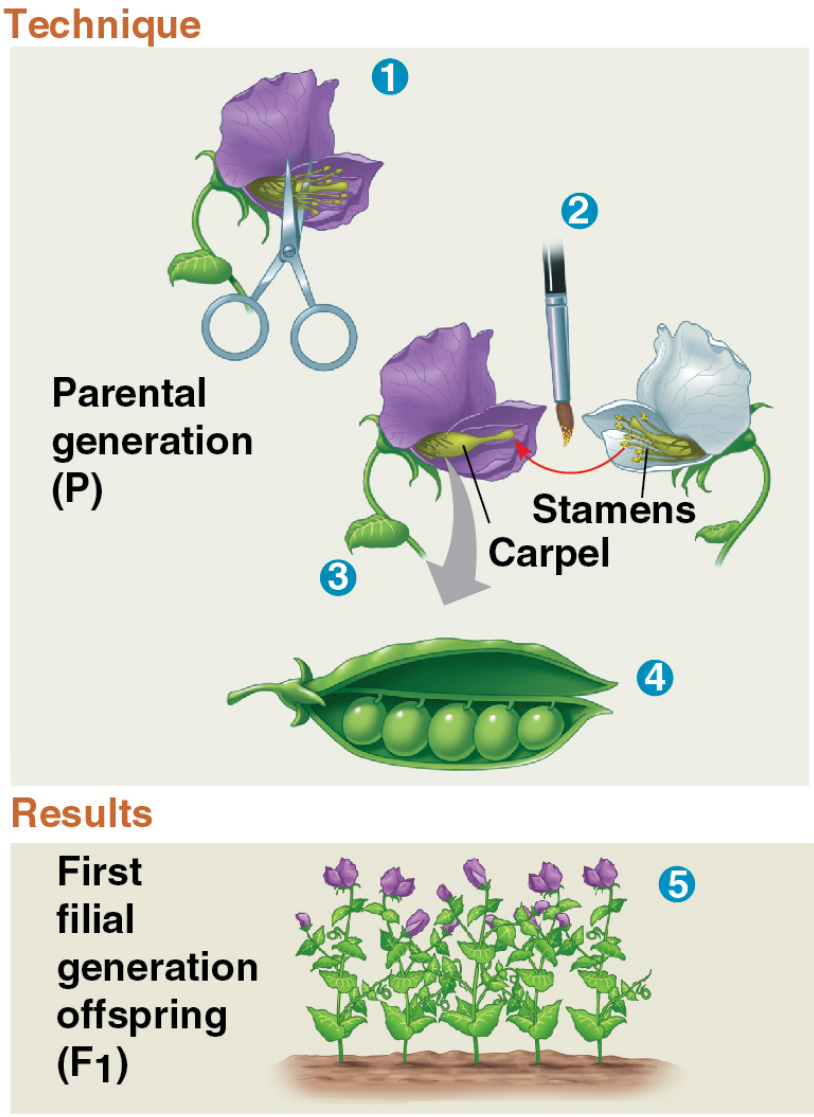
- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

Mendel's Experimental, Quantitative Approach

- Mendel's fresh approach to the study of heredity allowed him to deduce principles that had remained elusive to others
- A heritable feature that varies among individuals (such as flower color) is called a **character**
- Each variant for a character, such as purple or white color for flowers, is called a **trait**
- Peas were available to Mendel in many different varieties

- Other advantages of using peas
  - Short generation time
  - Large numbers of offspring
  - Mating could be controlled; plants could be allowed to self-pollinate or could be cross-pollinated

Figure 14.2


Technique
1
2
Parental generation (P)
Stamens
Carpel
3
4
Results
First filial generation offspring (F_1)
5

- Mendel chose to track only those characters that occurred in two distinct alternative forms
- He also started with varieties that were **true-breeding** (plants that produce offspring of the same variety when they self-pollinate)

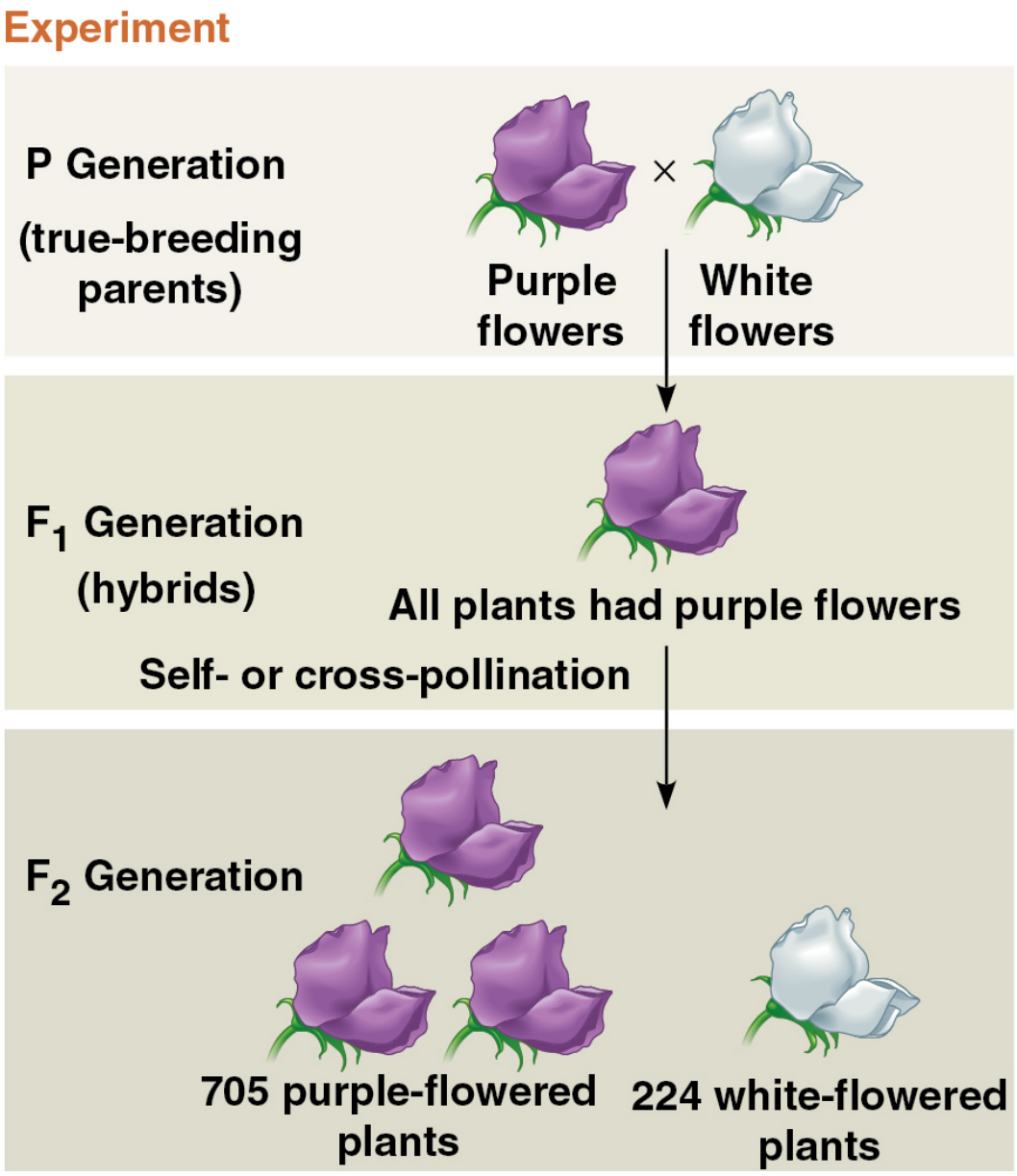
- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**
- The true-breeding parents are called the **P generation**
- The hybrid offspring of the P generation are called the **F_1 generation**
- When F_1 individuals self-pollinate or cross-pollinate with other F_1 hybrids, the **F_2 generation** is produced

The Law of Segregation

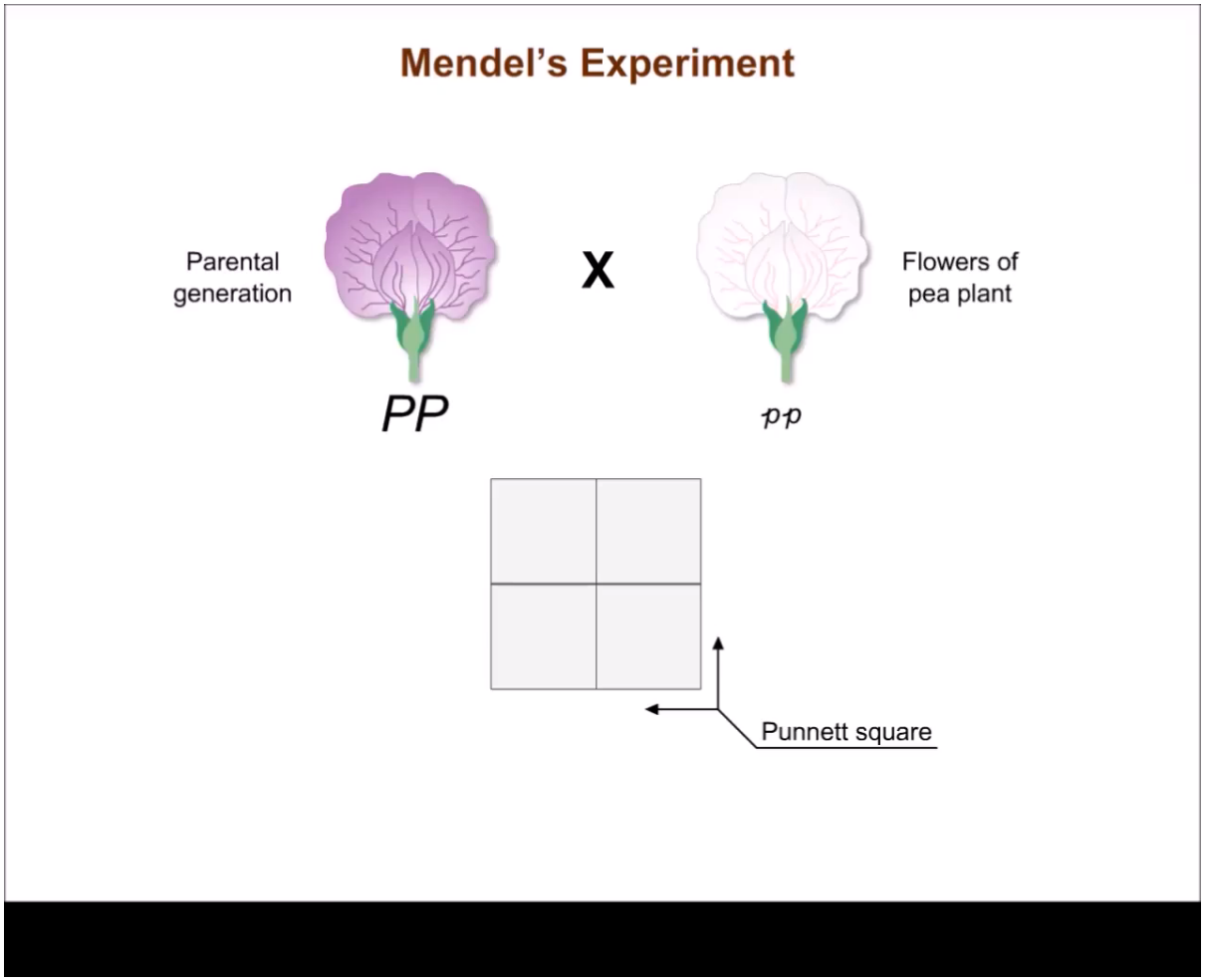
- In the 1800s, the explanation of heredity was the "blending" hypothesis
- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F_1 hybrids were purple
- This result was not predicted by the blending hypothesis

- 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 purple flowers to one white flower in the F_2 generation

Figure 14.3


Experiment
P Generation
(true-breeding parents)
Purple flowers
×
White flowers
F1 Generation
(hybrids)
All plants had purple flowers
Self- or cross-pollination
F2 Generation
705 purple-flowered plants
224 white-flowered plants

Video: Mendel's Cross on Flower Color



- 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
- The factor for white flowers was not diluted or destroyed because it reappeared in the F_2 generation

- 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

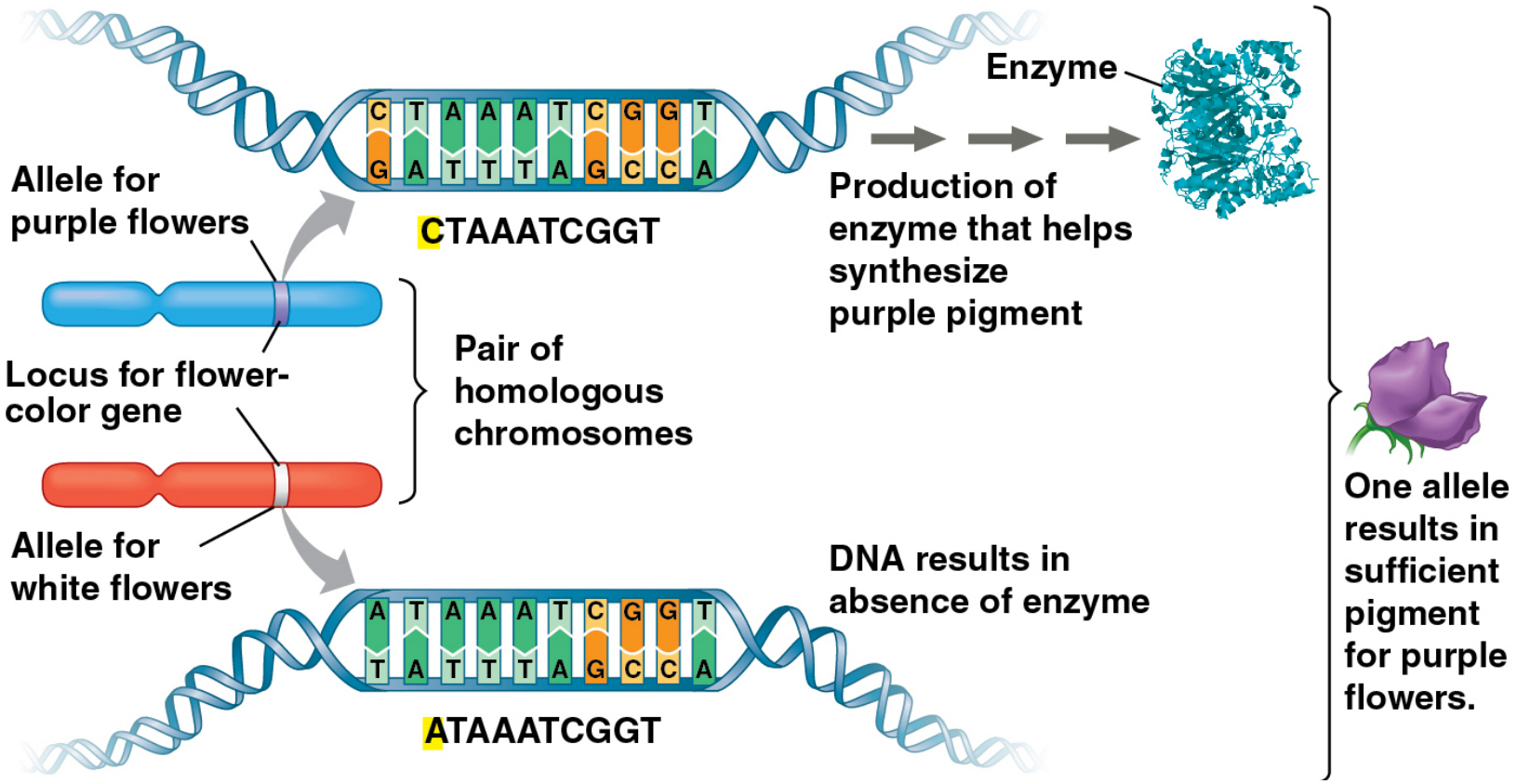
Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Pod shape	Inflated 	×	Constricted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

Mendel's Model

- Mendel developed a model 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

- First: 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 called **alleles**
- Each gene resides at a specific locus on a specific chromosome

Figure 14.4


Enzyme
C T A A A T C G G T
G A T T T A G C C A
CTAAATCGGT
Allele for purple flowers
Production of enzyme that helps synthesize purple pigment
Locus for flower-color gene
Pair of homologous chromosomes
Allele for white flowers
DNA results in absence of enzyme
A T A A A T C G G T
T A T T T A G C C A
ATAAATCGGT
One allele results in sufficient pigment for purple flowers.

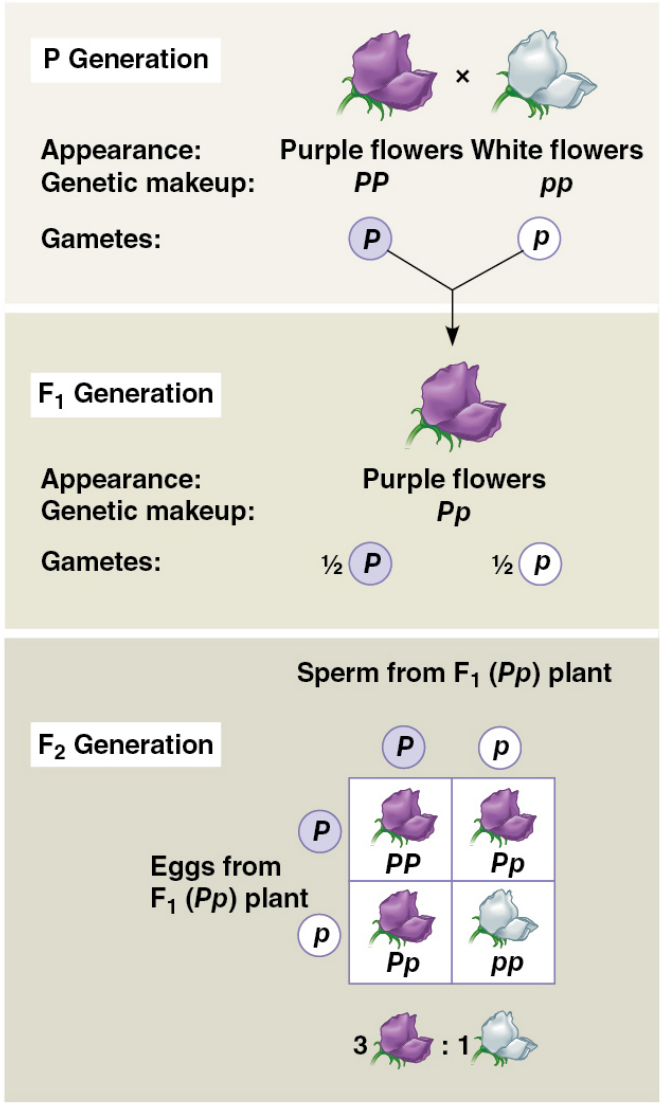
- Second: for each character, an organism inherits two alleles, one from each parent
- Mendel made this deduction without knowing about chromosomes
- The two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel's P generation
- Or the two alleles at a locus may differ, as in the F_1 hybrids

- Third: if the two alleles at a locus differ, then one, the **dominant allele**, determines the organism's appearance
- 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

- Fourth, the **law of segregation**: 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 organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

- The model accounts for the 3:1 ratio observed in the F_2 generation of Mendel's crosses
- Possible combinations of sperm and egg can be shown using a **Punnett square**
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele

Figure 14.5

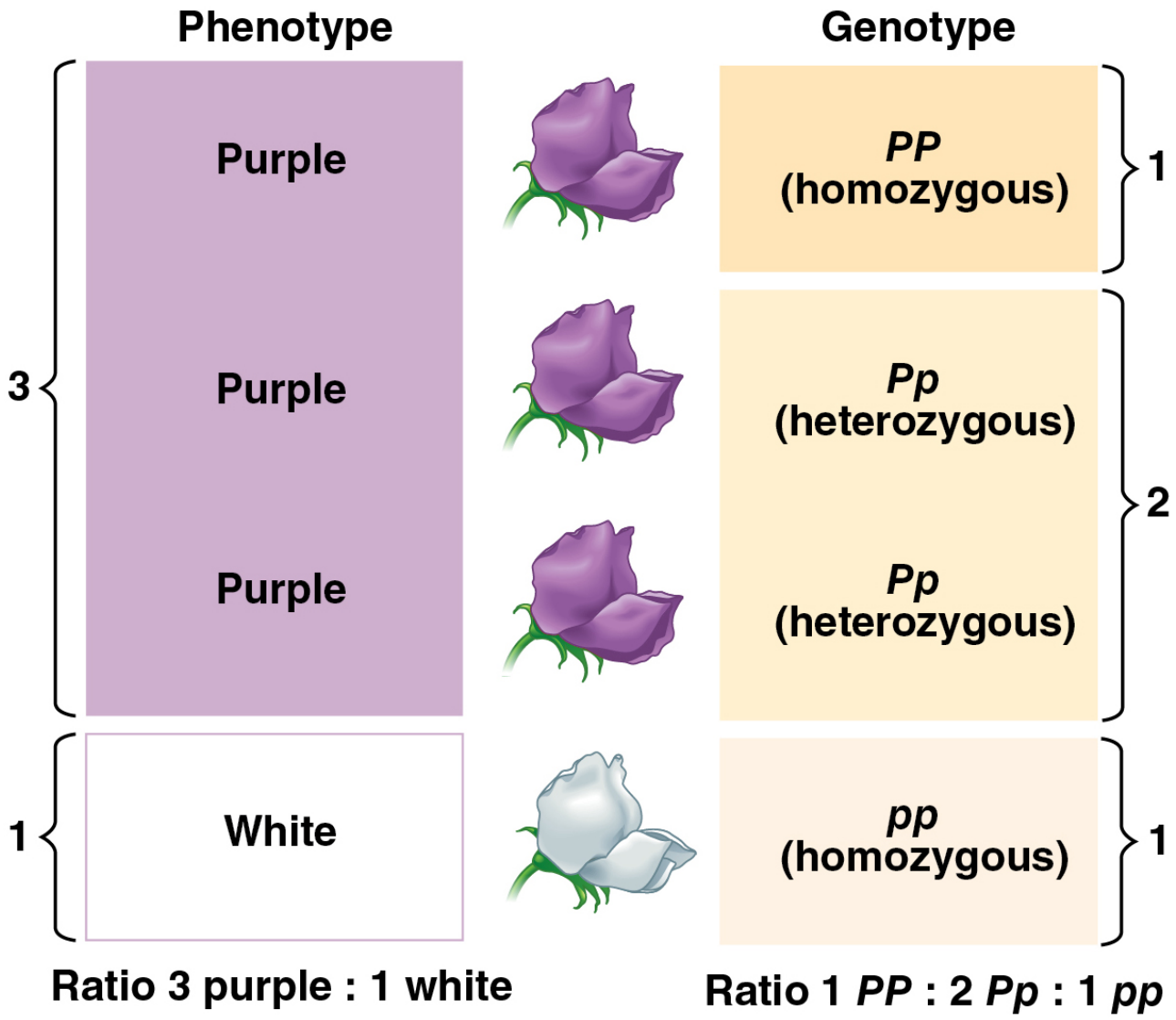

P Generation
×
Appearance:
Genetic makeup:
Purple flowers
PP
White flowers
pp
Gametes:
P
p
F1 Generation
Appearance:
Genetic makeup:
Purple flowers
Pp
Gametes:
½ P
½ p
Sperm from F1 (Pp) plant
F2 Generation
P
p
Eggs from F1 (Pp) plant
P
p
PP
Pp
Pp
pp
3 : 1

Useful Genetic Vocabulary

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

- 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

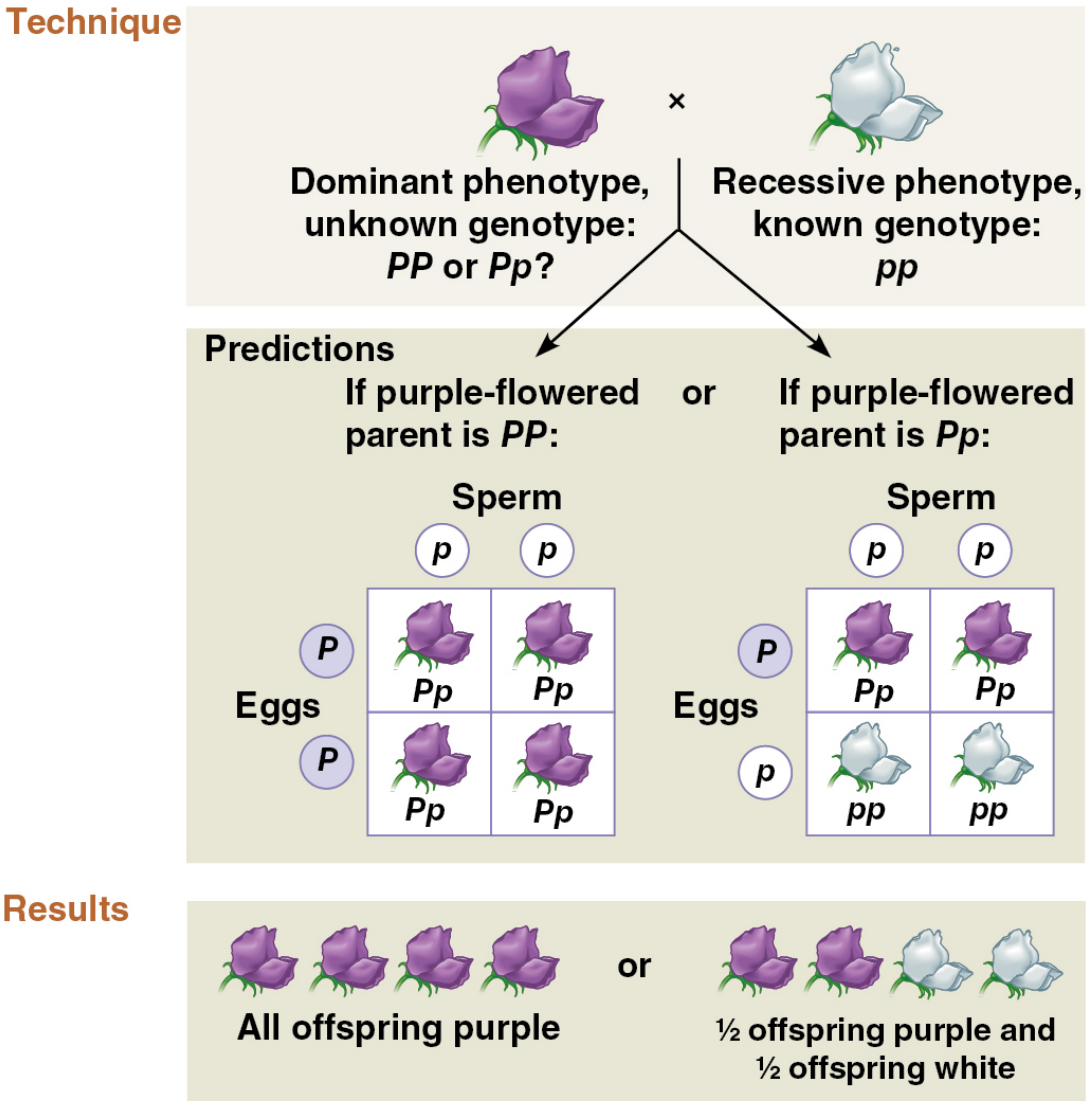
Figure 14.6


Phenotype
Genotype
Purple
Purple
Purple
White
3
1
PP (homozygous)
Pp (heterozygous)
Pp (heterozygous)
pp (homozygous)
1
2
1
Ratio 3 purple : 1 white
Ratio 1 PP : 2 Pp : 1 pp

The Testcross

- An individual with the dominant phenotype could be either homozygous dominant or heterozygous
- To determine the genotype we can 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

Figure 14.7

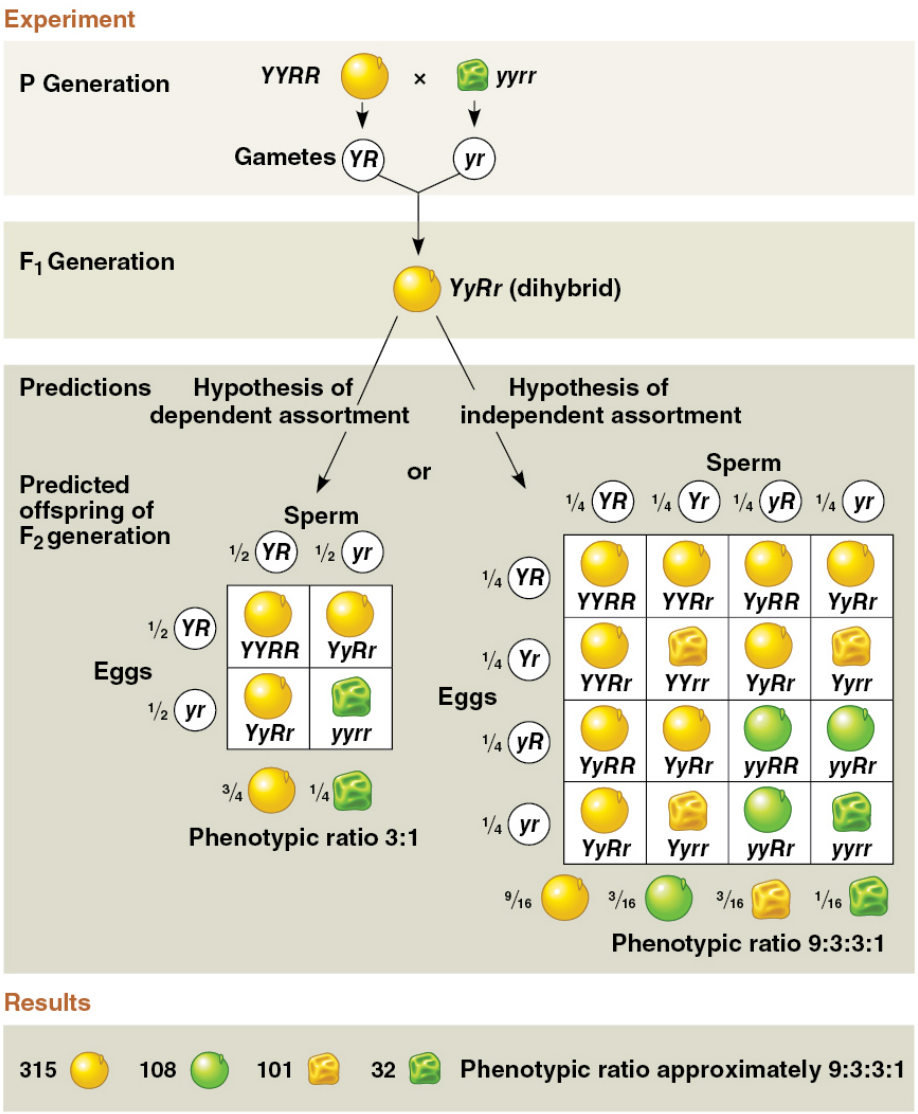

Technique
×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
Predictions
If purple-flowered parent is PP:
or
If purple-flowered parent is Pp:
Sperm
p
p
P
P
Eggs
Pp
Pp
Pp
Pp
Sperm
p
p
P
p
Eggs
Pp
Pp
pp
pp
Results
All offspring purple
or
½ offspring purple and ½ offspring 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**, meaning that they were 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 together as a unit or independently

Figure 14.8


Experiment
P Generation
YYRR
×
yyrr
Gametes
YR
yr
F1 Generation
YyRr (dihybrid)
Predictions
Hypothesis of dependent assortment
or
Hypothesis of independent assortment
Predicted offspring of F2 generation
Sperm
1/2 YR
1/2 yr
Eggs
1/2 YR
1/2 yr
YYRR
YyRr
YyRr
yyrr
3/4
1/4
Phenotypic ratio 3:1
Sperm
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
Eggs
1/4 YR
1/4 Yr
1/4 yR
1/4 yr
YYRR
YYRr
YyRR
YyRr
YYRr
YYrr
YyRr
Yyrr
YyRR
YyRr
yyRR
yyRr
YyRr
Yyrr
yyRr
yyrr
9/16
3/16
3/16
1/16
Phenotypic ratio 9:3:3:1
Results
315
108
101
32
Phenotypic ratio approximately 9:3:3:1

- Using a dihybrid cross, Mendel developed the **law of independent assortment**
- It states that each pair of alleles segregates independently of any other pair of alleles during gamete formation
- This law applies only to genes on different, nonhomologous chromosomes or those far apart on the same chromosome
- Genes located near each other on the same chromosome tend to be inherited together

CONCEPT 14.2: Probability laws govern Mendelian inheritance

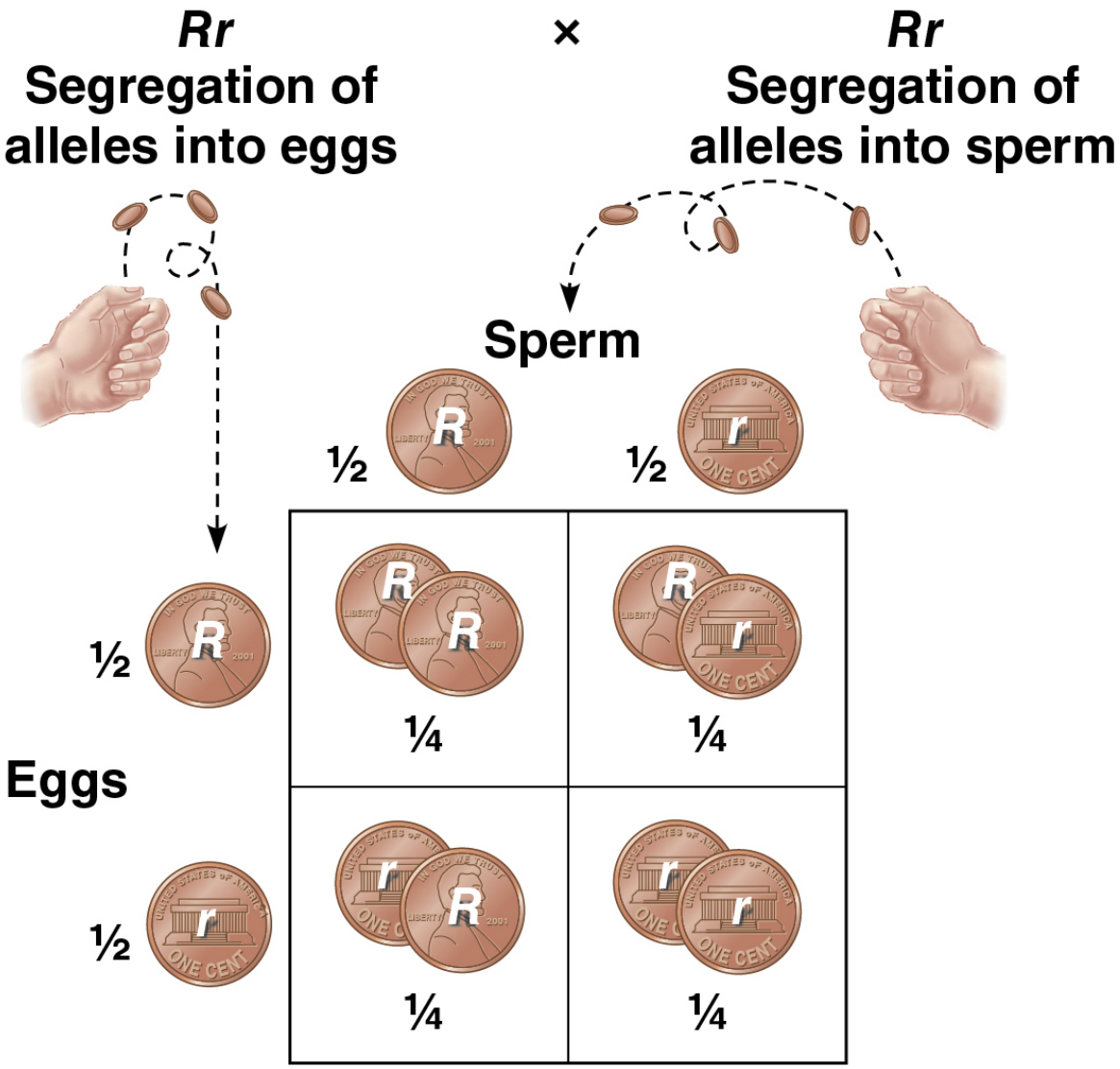
- Mendel's laws of segregation and independent assortment reflect the rules of probability that apply to tossing coins or rolling dice
- When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss
- In the same way, the alleles of one gene segregate into gametes independently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

- The **addition rule** states that the probability that any one of two or more mutually 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

Figure 14.9


Rr
Segregation of alleles into eggs
×
Rr
Segregation of alleles into sperm
Sperm
½
½
Eggs
½
½
¼
¼
¼
¼

Solving Complex Genetics Problems with the Rules of Probability

- We can apply the rules of probability to predict the outcome of crosses involving multiple characters
- A 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

Figure 14.UN01

Probability of *YYRR* = ¼ (probability of *YY*) × ¼ (*RR*) = 1/16

Probability of *YyRR* = ½ (*Yy*) × ¼ (*RR*) = 1/8

Figure 14.UN02

ppyyRr	¼ (probability of *pp*)	× ½ (*yy*)	× ½ (*Rr*)	= 1/16
ppYyrr	¼ (*pp*)	× ½ (*Yy*)	× ½ (*rr*)	= 1/16
Ppyyrr	½ (*Pp*)	× ½ (*yy*)	× ½ (*rr*)	= 2/16
PPyyrr	¼ (*PP*)	× ½ (*yy*)	× ½ (*rr*)	= 1/16
ppyyrr	¼ (*pp*)	× ½ (*yy*)	× ½ (*rr*)	= 1/16

Chance of *at least two* recessive traits = 6/16 = 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

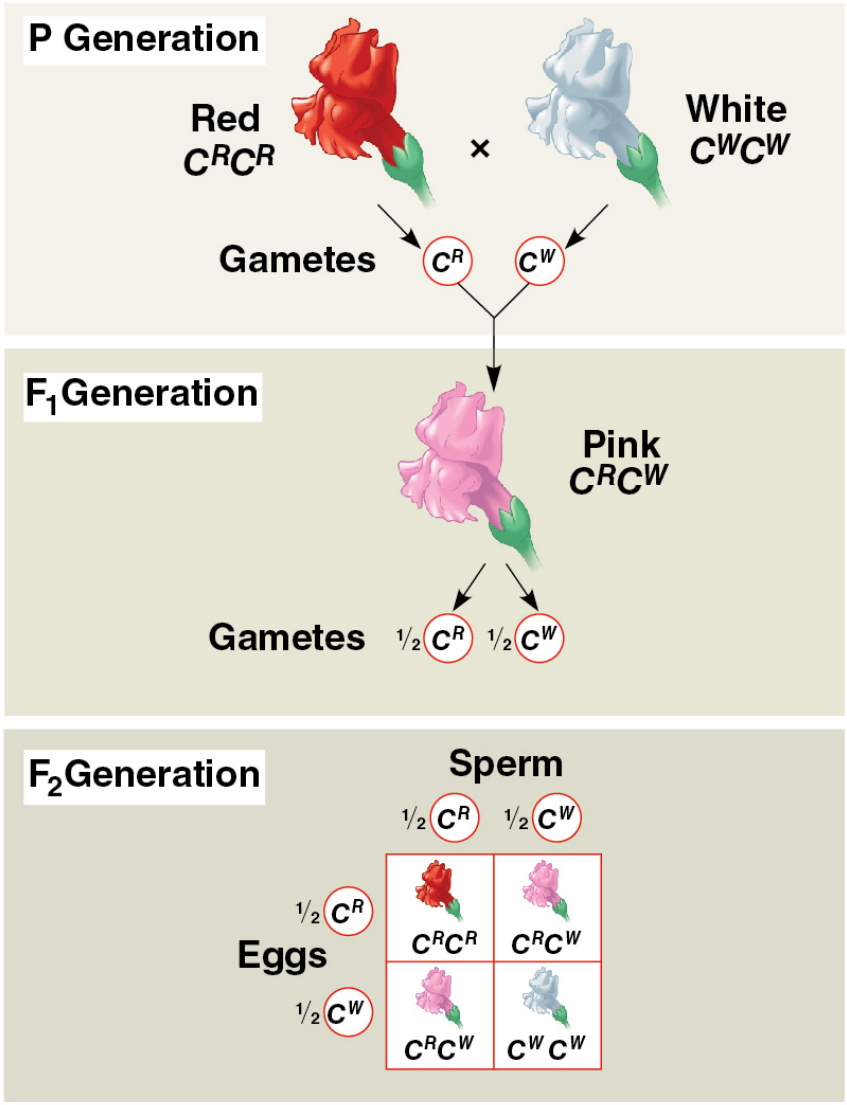
Extending Mendelian Genetics for a Single Gene

- 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

Figure 14.10


P Generation
Red
C^RC^R
×
White
C^WC^W
Gametes
C^R
C^W
F_1 Generation
Pink
C^RC^W
Gametes
½ C^R
½ C^W
F_2 Generation
Sperm
½ C^R
½ C^W
Eggs
½ C^R
½ C^W
C^RC^R
C^RC^W
C^RC^W
C^WC^W

The Relationship Between Dominance and Phenotype

- In the case of pea shape, the dominant allele codes for an enzyme that converts an unbranched form of starch in the seed to a branched form
- The recessive allele codes for a defective form of the enzyme, which leads to an accumulation of unbranched starch
- This causes water to enter the seed, which then wrinkles as it dries

- **Tay-Sachs disease** is a fatal inherited disorder; 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 (that is, the enzyme activity level) is incompletely dominant
  - At the molecular level, the alleles are codominant

Frequency of Dominant Alleles

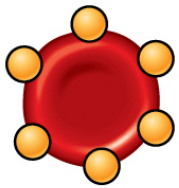
- Dominant alleles are not necessarily more common in populations than recessive alleles
- One baby out of 400 in the United States is born with extra fingers or toes
- This condition, polydactyly, is caused by a dominant allele, found much less frequently in the population than the recessive allele

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 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

Figure 14.11

(a) The three alleles for the ABO blood groups and their carbohydrates			
Allele	I^A	I^B	i
Carbohydrate	A 	B 	none

(b) Blood group genotypes and phenotypes				
Genotype	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell with surface carbohydrates				
Phenotype (blood group)	A	B	AB	O

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

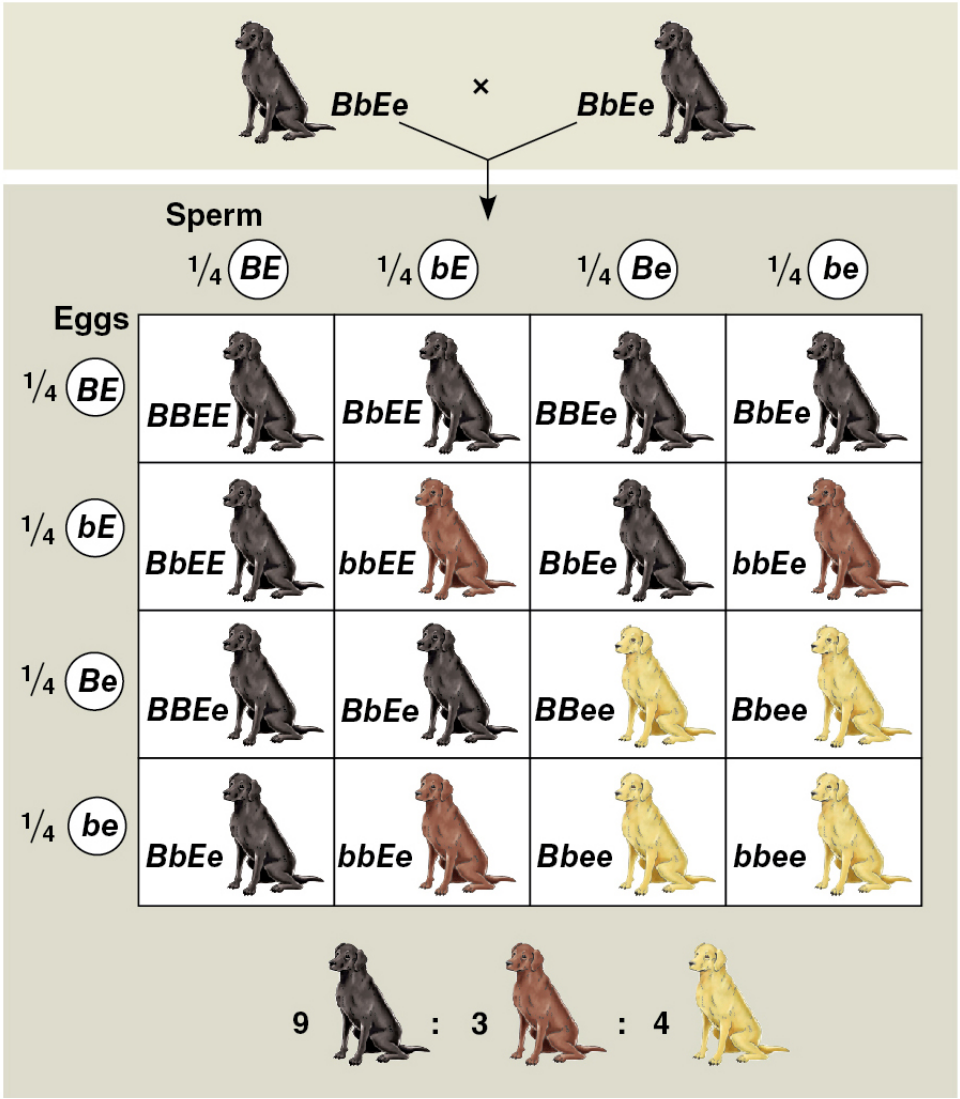
- Some traits may be determined by two or more genes
- In epistasis, one gene affects the phenotype of another due to interaction of their gene products
- In polygenic inheritance, multiple genes independently affect a single trait

Epistasis

- In **epistasis**, expression of a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers 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 *E* for color and *e* for no color) determines whether the pigment will be deposited in the hair

- If heterozygous black labs (genotype *BbEe*) are mated, we might expect the dihybrid F_2 ratio of 9:3:3:1
- However, a Punnett square shows that the phenotypic ratio will be 9 black to 3 chocolate to 4 yellow labs
- Epistatic interactions produce a variety of ratios, all of which are modified versions of 9:3:3:1

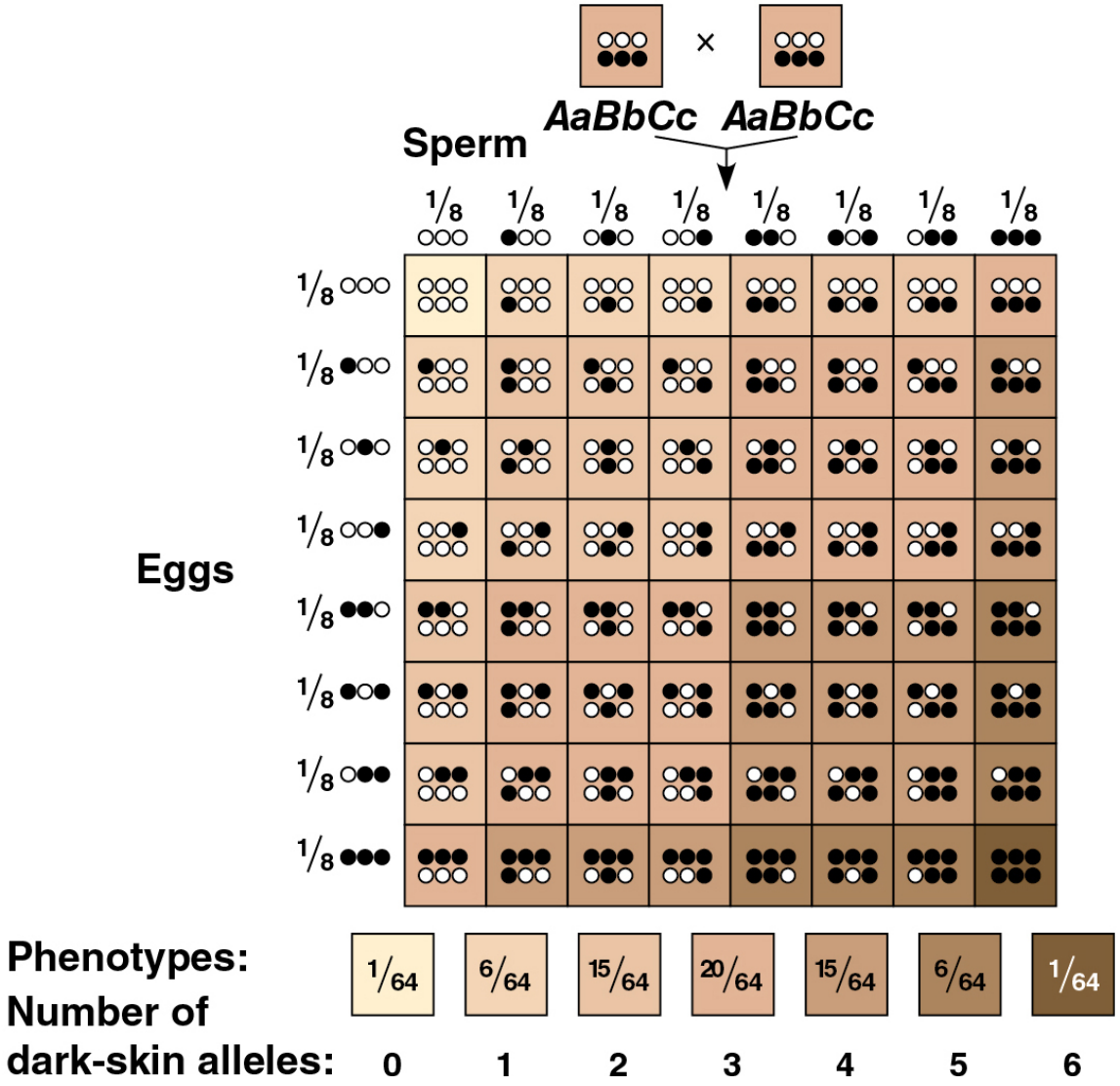
Figure 14.12


BbEe
×
BbEe
Sperm
1/4 BE
1/4 bE
1/4 Be
1/4 be
Eggs
1/4 BE
1/4 bE
1/4 Be
1/4 be
BBEE
BbEE
BBEe
BbEe
BbEE
bbEE
BbEe
bbEe
BBEe
BbEe
BBee
Bbee
BbEe
bbEe
Bbee
bbee
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
- Height is a good example of polygenic inheritance; over 180 genes affect height
- Skin pigmentation in humans is also controlled by many separately inherited genes

Figure 14.13


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

Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from simple Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype
- The phenotypic range is broadest for polygenic characters
- Traits that depend on multiple genes combined with environmental influences are called **multifactorial**

Figure 14.14

(a) Hydrangeas grown in basic soil

(b) Hydrangeas of the same genetic variety grown in acidic soil with free aluminum

A Mendelian View of Heredity and Variation

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

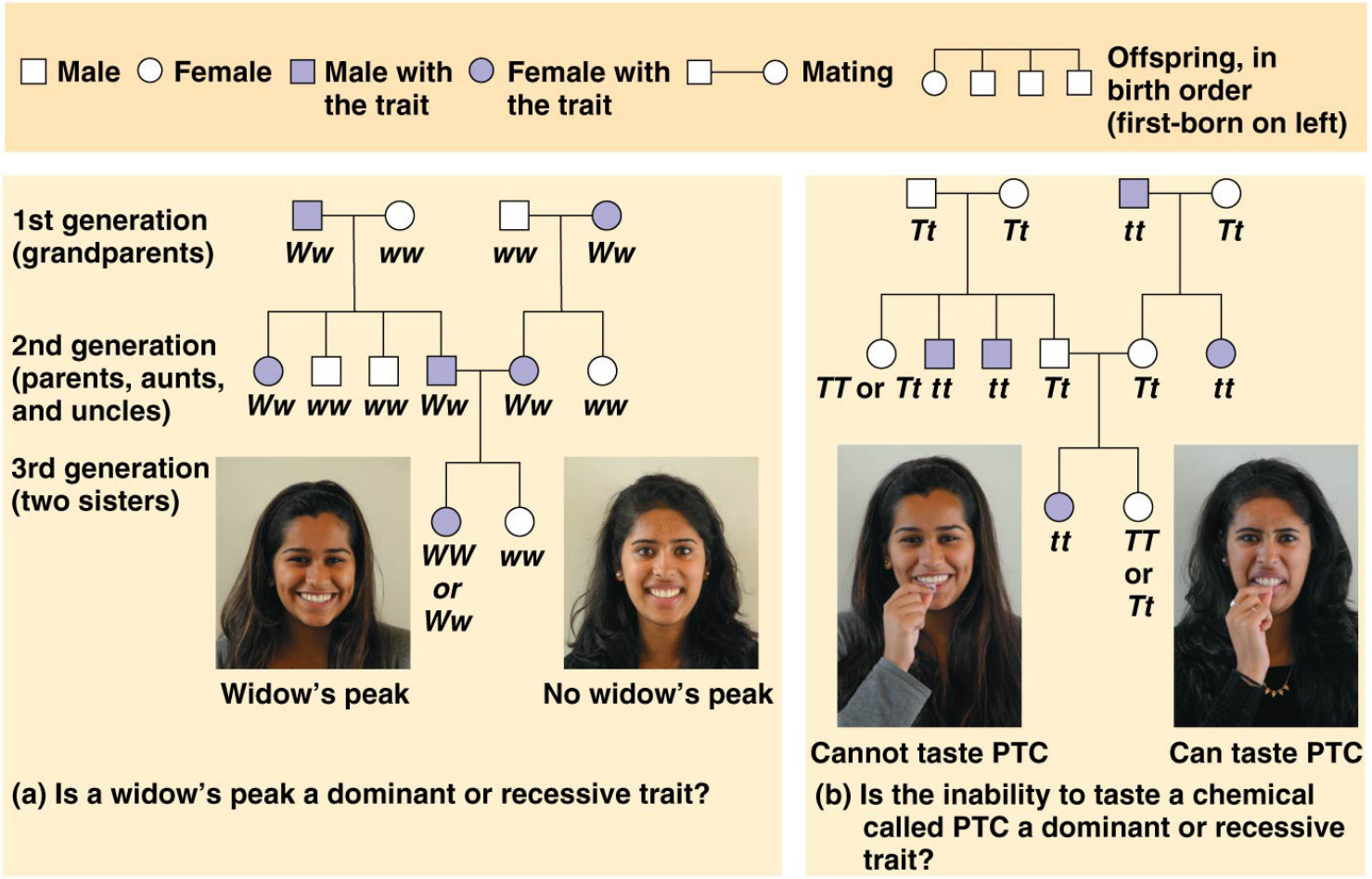
CONCEPT 14.4: Many Human Traits follow Mendelian Patterns of Inheritance

- 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

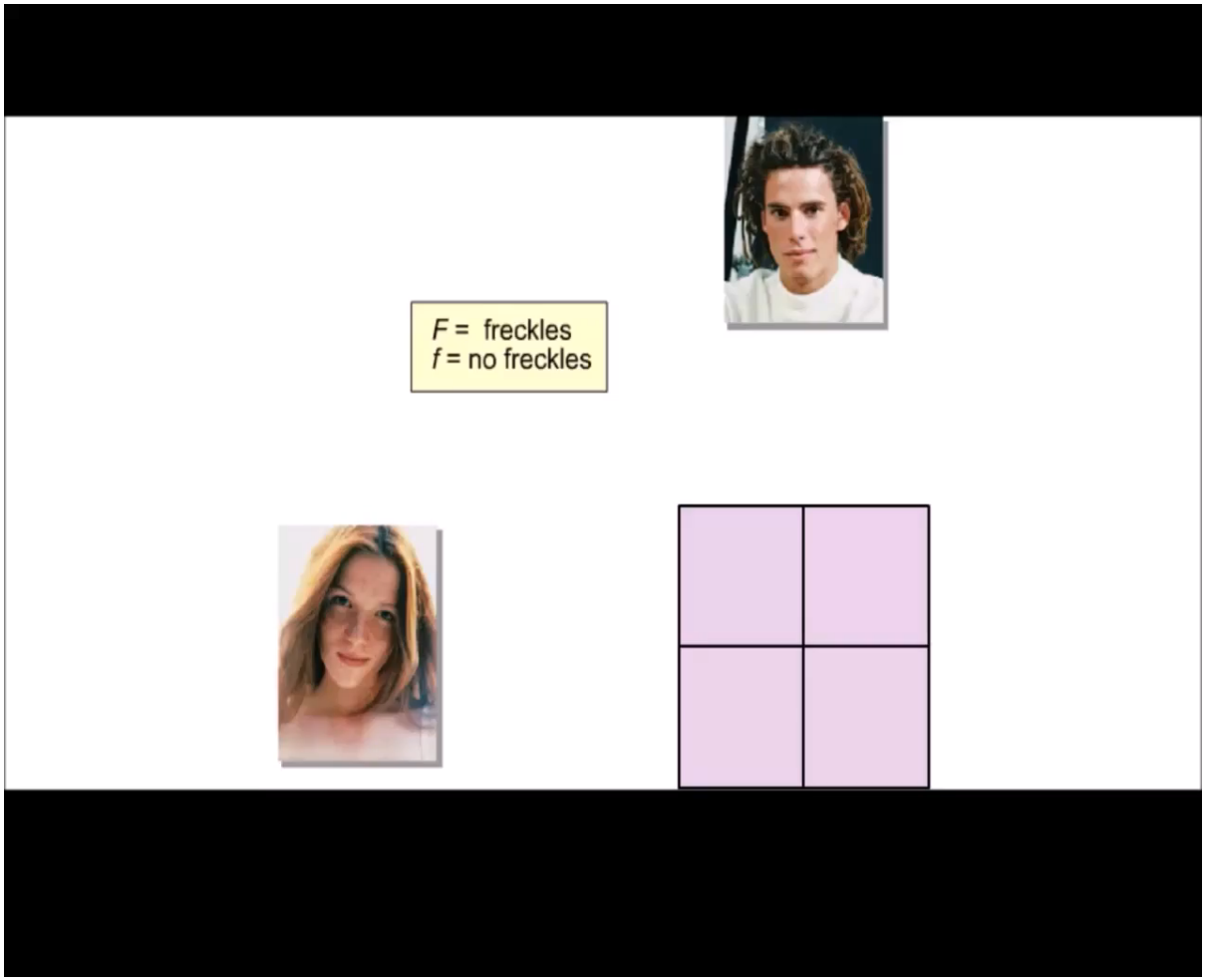
Pedigree Analysis

- In human genetics, geneticists analyze the results of human matings that have already occurred
- A **pedigree** is a family tree that describes the inheritance of a trait across generations

Figure 14.15


Male
Female
Male with the trait
Female with the trait
Mating
Offspring, in birth order (first-born on left)
1st generation (grandparents)
2nd generation (parents, aunts, and uncles)
3rd generation (two sisters)
Ww
ww
ww
Ww
Ww ww ww Ww Ww ww
WW or Ww
ww
Widow's peak
No widow's peak
(a) Is a widow's peak a dominant or recessive trait?
Tt
Tt
tt
Tt
TT or Tt
tt
tt
Tt
Tt
tt
tt
TT or Tt
Cannot taste PTC
Can taste PTC
(b) Is the inability to taste a chemical called PTC a dominant or recessive trait?

Animation: Simplified Cross of One Character in Humans



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

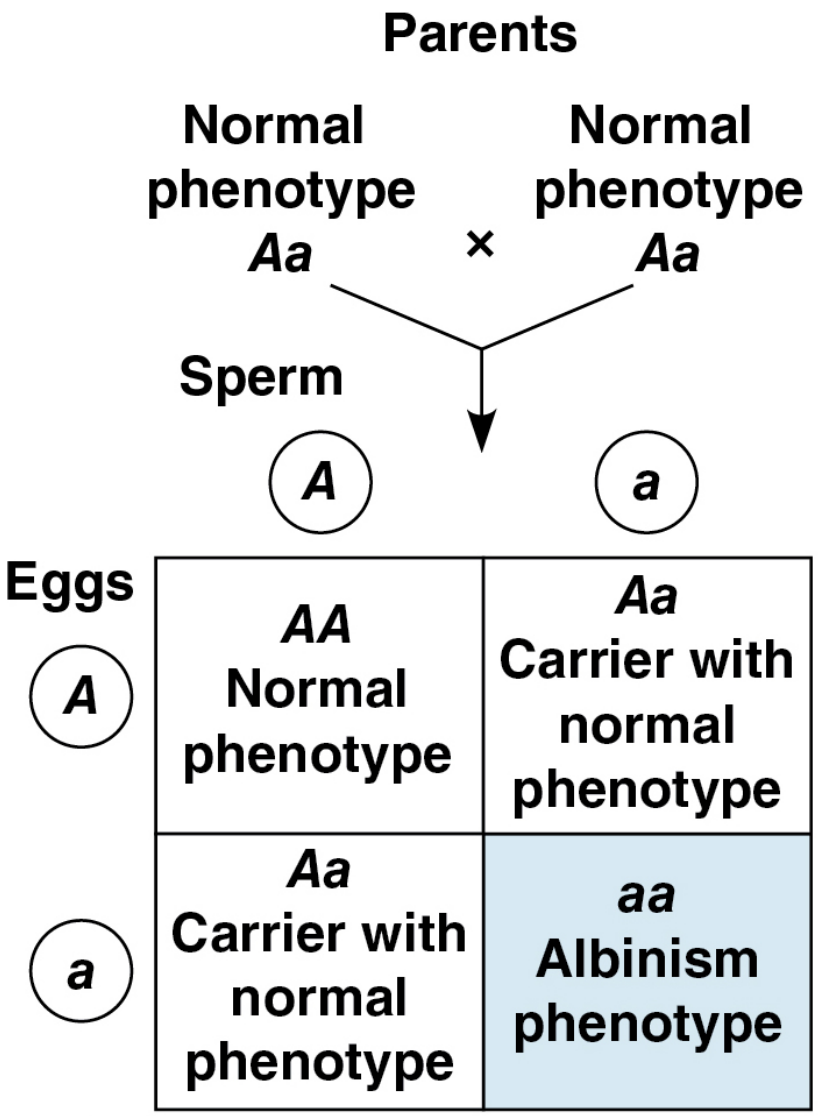
Recessively Inherited Disorders

- Many genetic disorders are inherited in a recessive manner
- These range from relatively mild to life-threatening

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
- Most individuals with recessive disorders are born to carrier parents
- Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair

Figure 14.16


Parents
Normal phenotype
Aa
×
Normal phenotype
Aa
Sperm
A
a
Eggs
A
a
AA
Normal phenotype
Aa
Carrier with normal phenotype
Aa
Carrier with normal phenotype
aa
Albinism phenotype

- If a recessive allele that causes a disease is rare, it is unlikely that two carriers will meet and mate
- Consanguineous matings (that is, between close relatives) increase the chance that both parents of a child carry 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, leading to a buildup of chloride ions outside the cell
- Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine

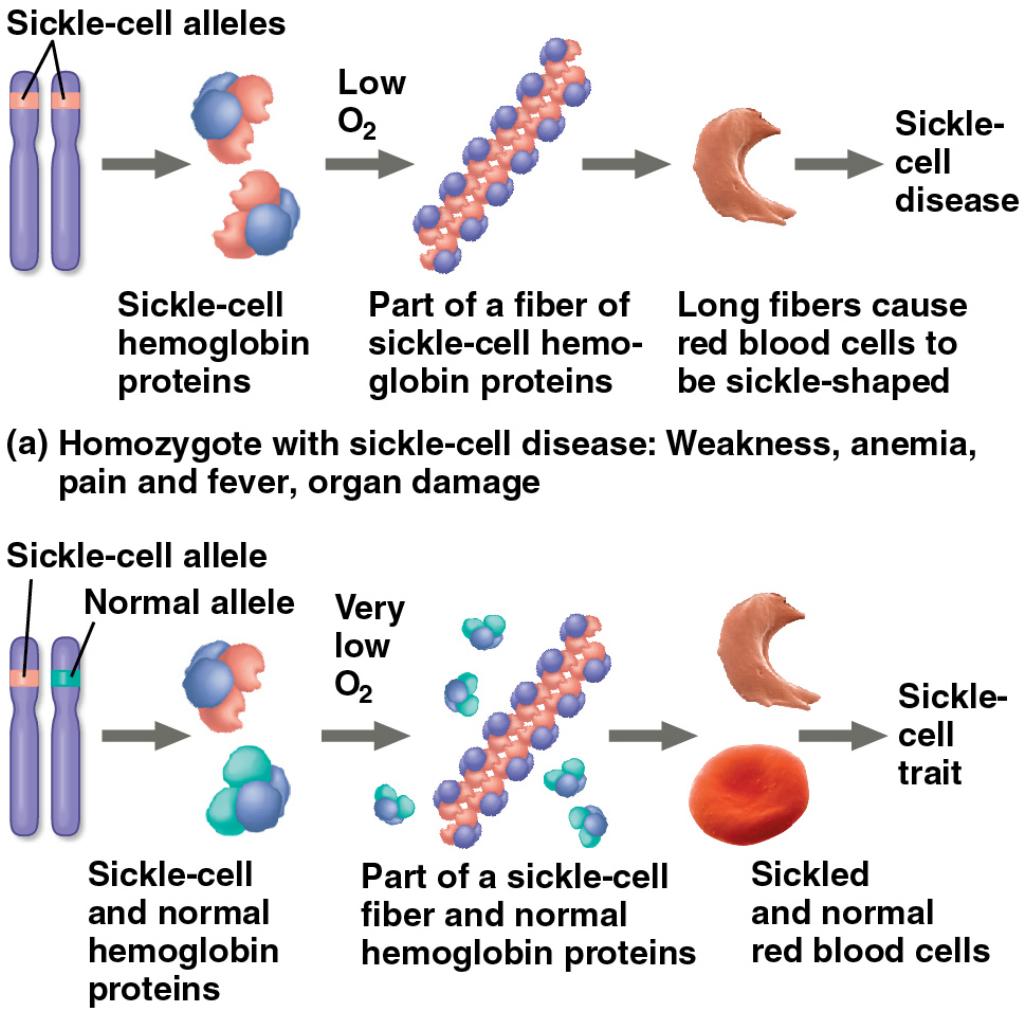
- Untreated, cystic fibrosis can cause death by the age of 5
- Daily doses of antibiotics to stop infection and physical therapies can prolong life
- In the United States, more than half of those with cystic fibrosis now survive into their 40s

Sickle-Cell Disease: A Genetic Disorder with Evolutionary Implications

- **Sickle-cell disease** affects one out of 400 African-Americans
- It is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- In homozygous individuals, all hemoglobin is abnormal (sickle-cell)
- Symptoms include physical weakness, pain, organ damage, and even paralysis

- Heterozygotes (said to have sickle-cell trait) are usually healthy but may suffer some symptoms
- About one out of ten African-Americans has sickle-cell trait, an unusually high frequency
- Heterozygotes are less susceptible to the malaria parasite, so there is an advantage to being heterozygous in regions where malaria is common

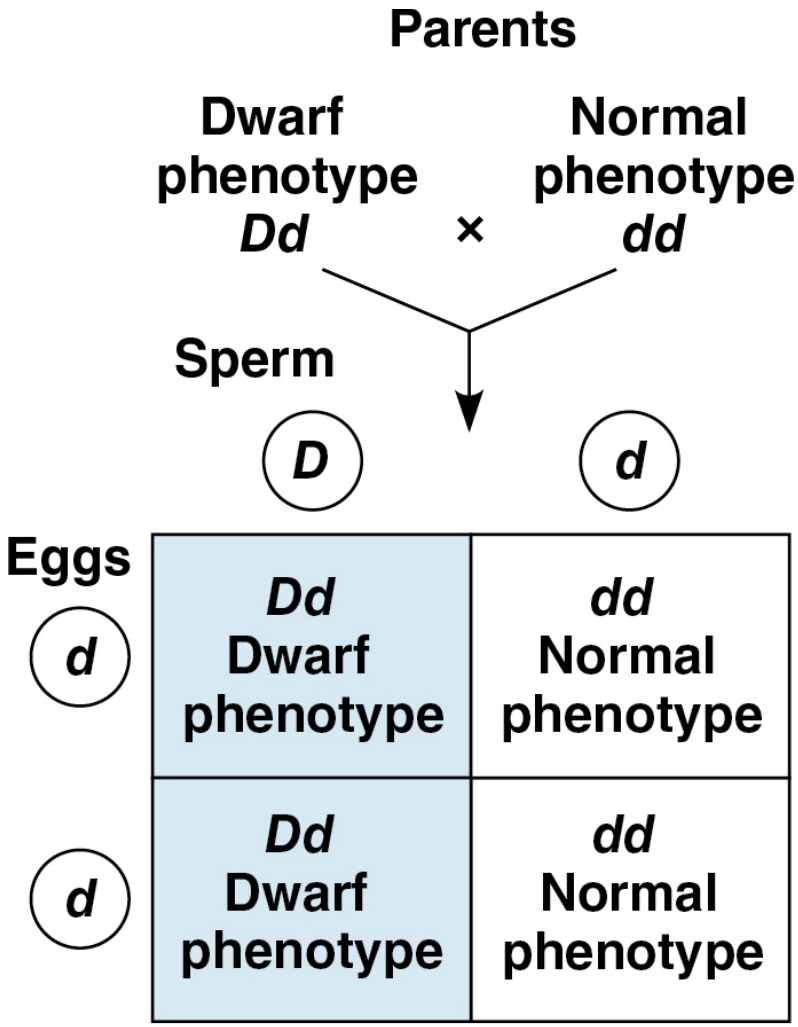
Figure 14.17

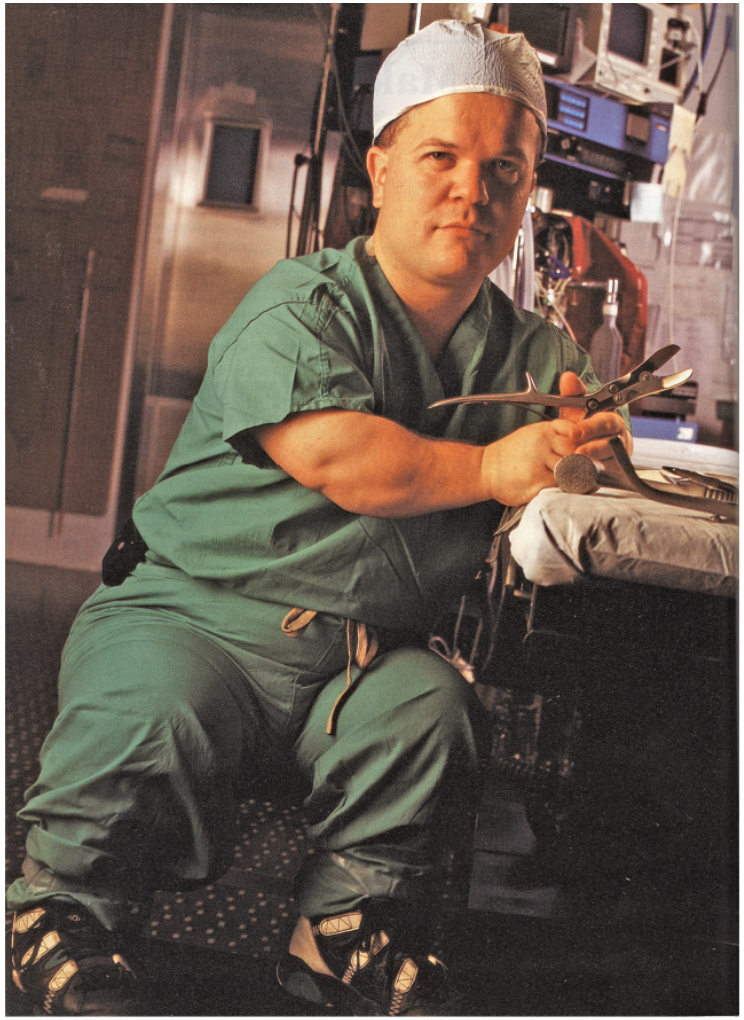

Sickle-cell alleles
Low O_2
Sickle-cell disease
Sickle-cell hemoglobin proteins
Part of a fiber of sickle-cell hemo-globin proteins
Long fibers cause red blood cells to be sickle-shaped
(a) Homozygote with sickle-cell disease: Weakness, anemia, pain and fever, organ damage
Sickle-cell allele
Normal allele
Very low O_2
Sickle-cell trait
Sickle-cell and normal hemoglobin proteins
Part of a sickle-cell fiber and normal hemoglobin proteins
Sickled and normal red blood cells
(b) Heterozygote with sickle-cell trait: Some symptoms when blood oxygen is very low; reduction of malaria symptoms

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

Figure 14.18


Parents
Dwarf phenotype Dd
×
Normal phenotype dd
Sperm
D
d
Eggs
d
d
Dd Dwarf phenotype
dd Normal phenotype
Dd Dwarf phenotype
dd Normal phenotype



- The timing of onset of a disease significantly affects its inheritance
- **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
- Once the deterioration of the nervous system begins, the condition is irreversible and fatal

- There is a test that can detect the presence of the Huntington's allele in an individual's genome
- Some individuals with a family history of Huntington's disease choose to be tested for the allele
- Others decide that it would be too stressful to find out

Multifactorial Disorders

- Many diseases, such as heart disease, cancer, alcoholism, and mental illnesses, have both genetic and environmental components
- No matter what our genotype, our lifestyle has a tremendous effect on phenotype

Genetic Testing and Counseling

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease
- Fetal and newborn testing can also reveal genetic disorders

Counseling Based on Mendelian Genetics and Probability Rules

- Suppose a couple both have a brother who died from the same recessively inherited disease
- A genetic counselor can help determine the risk that this couple will have a child with the disease
- It is important to remember that each child represents an independent event in the sense that its genotype is unaffected by the genotypes of older siblings

- If both members of the couple had a sibling with the recessively inherited illness, both of their parents were carriers
- Thus each has a ⅔ chance of being a carrier themselves
- If both are carriers, there is a ¼ chance of each child having the recessive illness
- The overall probability of them having a child with the illness is ⅔ × ⅔ × ¼ = 1/9

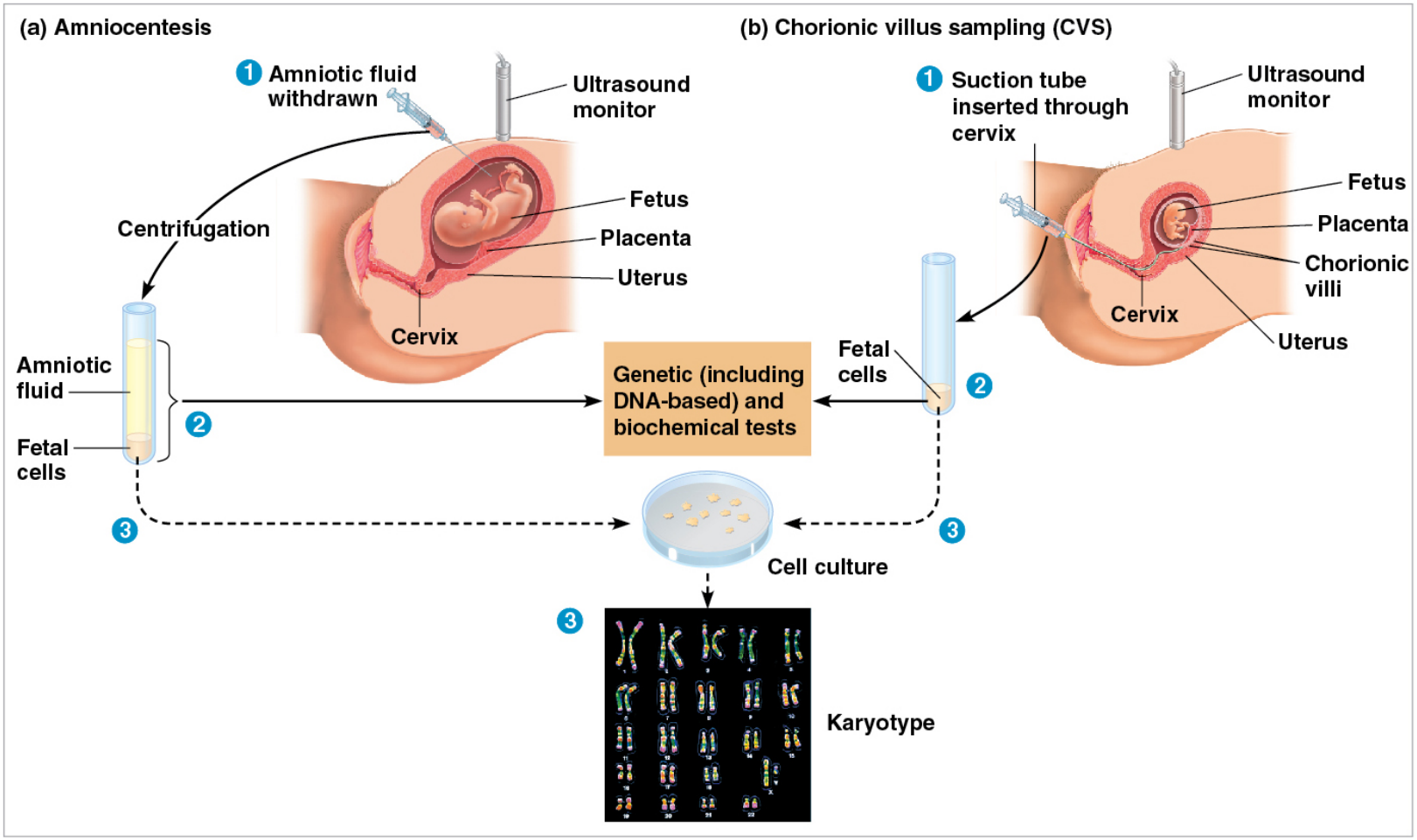
Tests for Identifying Carriers

- For a growing number of diseases, tests are available that identify carriers and help define the odds of having an affected child more accurately
- The tests enable people to make more informed decisions about having children
- However, they raise other issues, such as whether affected individuals fully understand their genetic test results, and how the test results are used

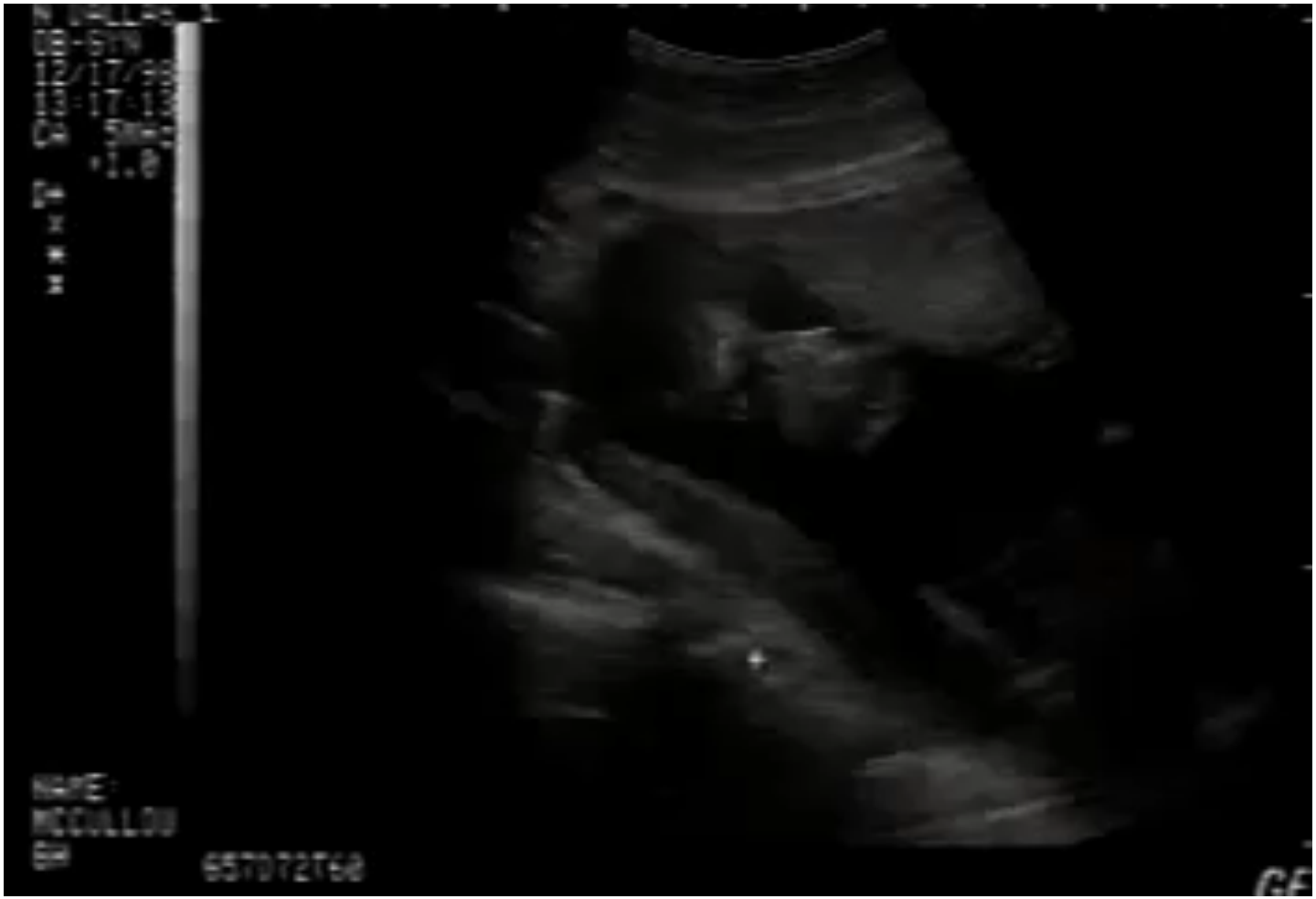
Fetal Testing

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

Figure 14.19

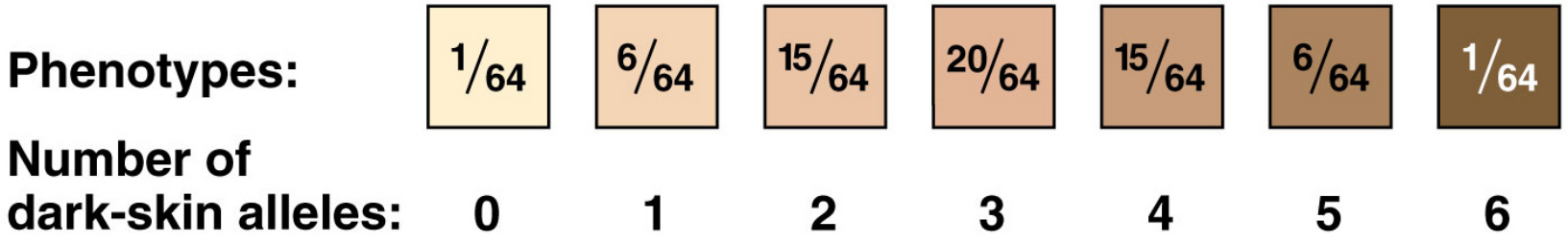

(a) Amniocentesis
1 Amniotic fluid withdrawn
Ultrasound monitor
Fetus
Placenta
Uterus
Cervix
Centrifugation
Amniotic fluid
Fetal cells
2
3
(b) Chorionic villus sampling (CVS)
1 Suction tube inserted through cervix
Ultrasound monitor
Fetus
Placenta
Chorionic villi
Cervix
Uterus
Fetal cells
2
3
Genetic (including DNA-based) and biochemical tests
Cell culture
3
Karyotype

Video: Ultra Sound of Fetus



Newborn Screening

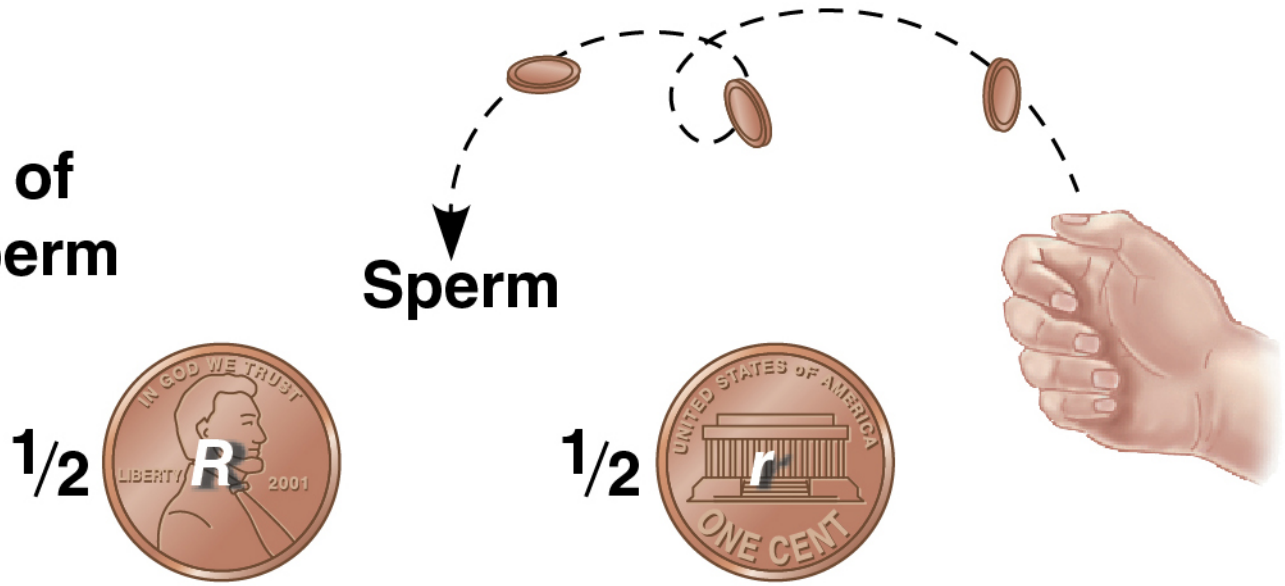
- Some genetic disorders can be detected at birth by simple tests that are now routinely performed in most hospitals in the United States
- One common test is for phenylketonuria (PKU), a recessively inherited disorder that occurs in one of every 10,000–15,000 births in the United States
- The number of conditions that can be tested in newborns is over 100 and ever-increasing

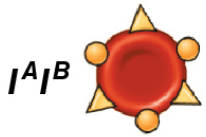


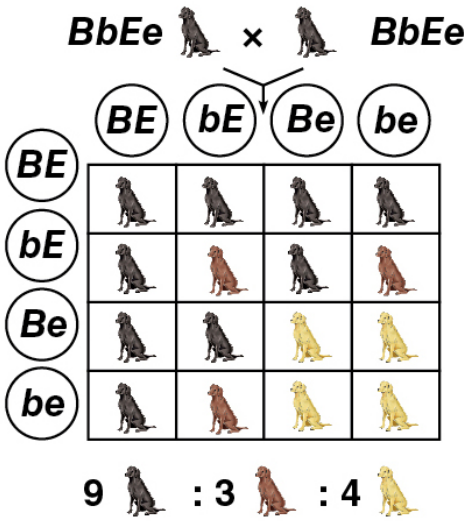
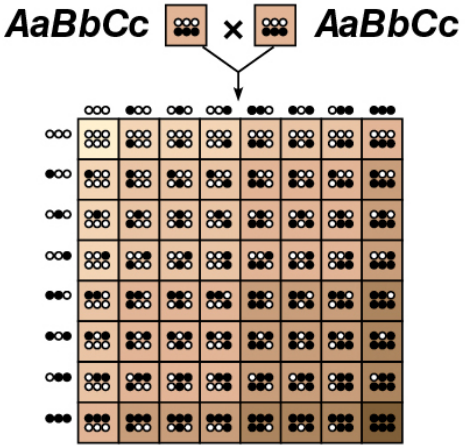
Further Reading R. A. Sturm, A golden age of human pigmentation genetics, *Trends in Genetics* 22:464–468 (2006).

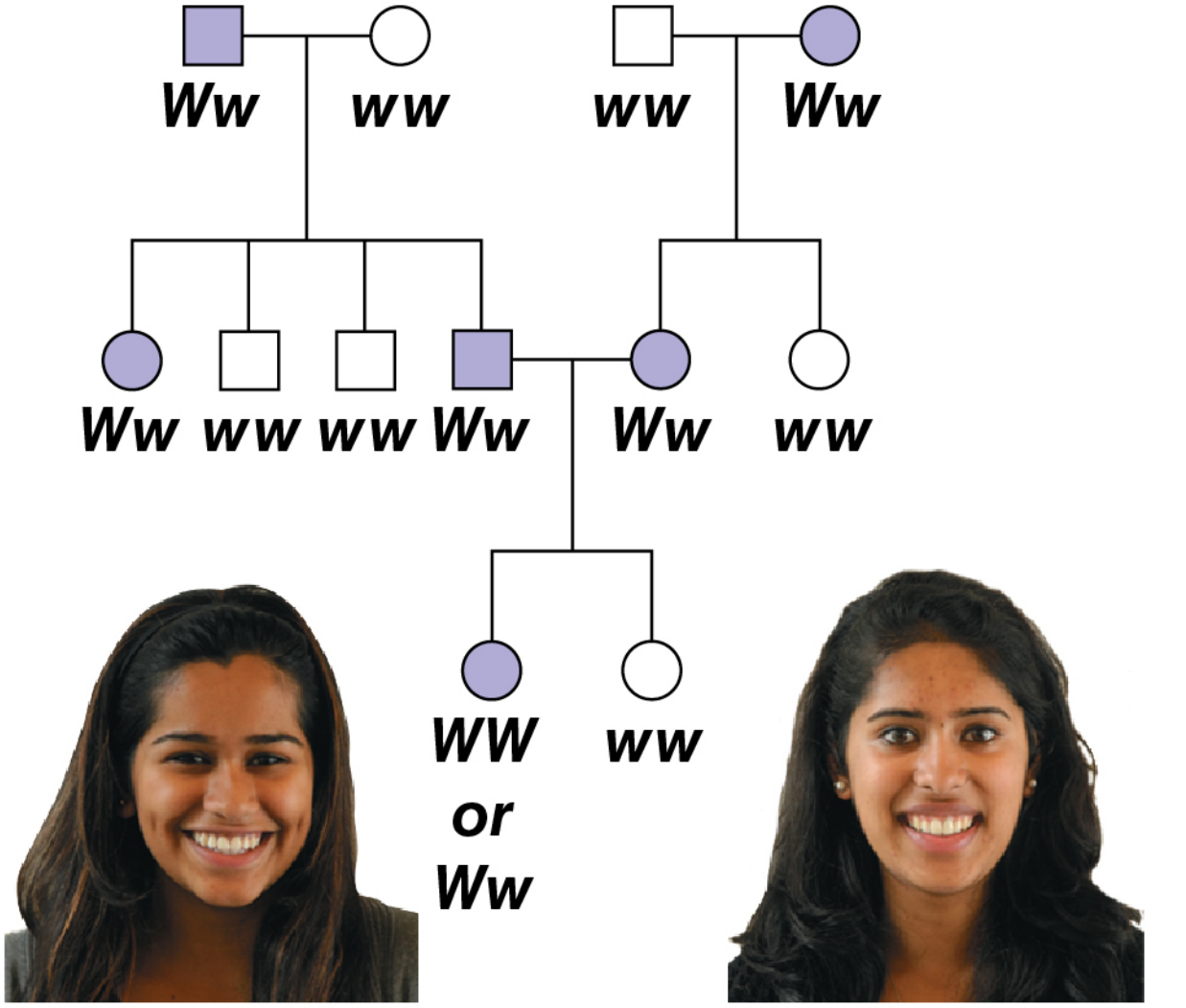
Rr

Segregation of alleles into sperm



Relationship among alleles of a single gene	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	Both phenotypes expressed in heterozygotes	
Multiple alleles	In the population, some genes have more than two alleles	ABO blood group alleles I^A, I^B, i
Pleiotropy	One gene affects multiple phenotypic characters	Sickle-cell disease

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


Ww
ww
ww
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ww
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WW
or
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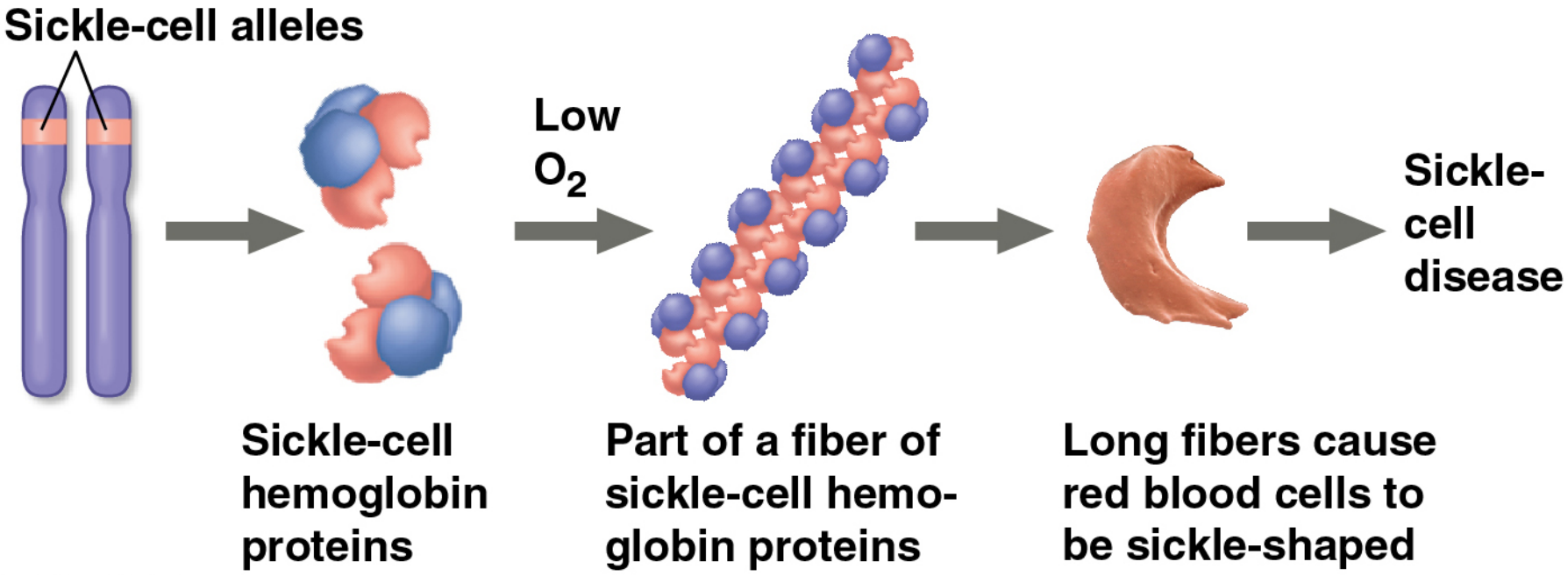
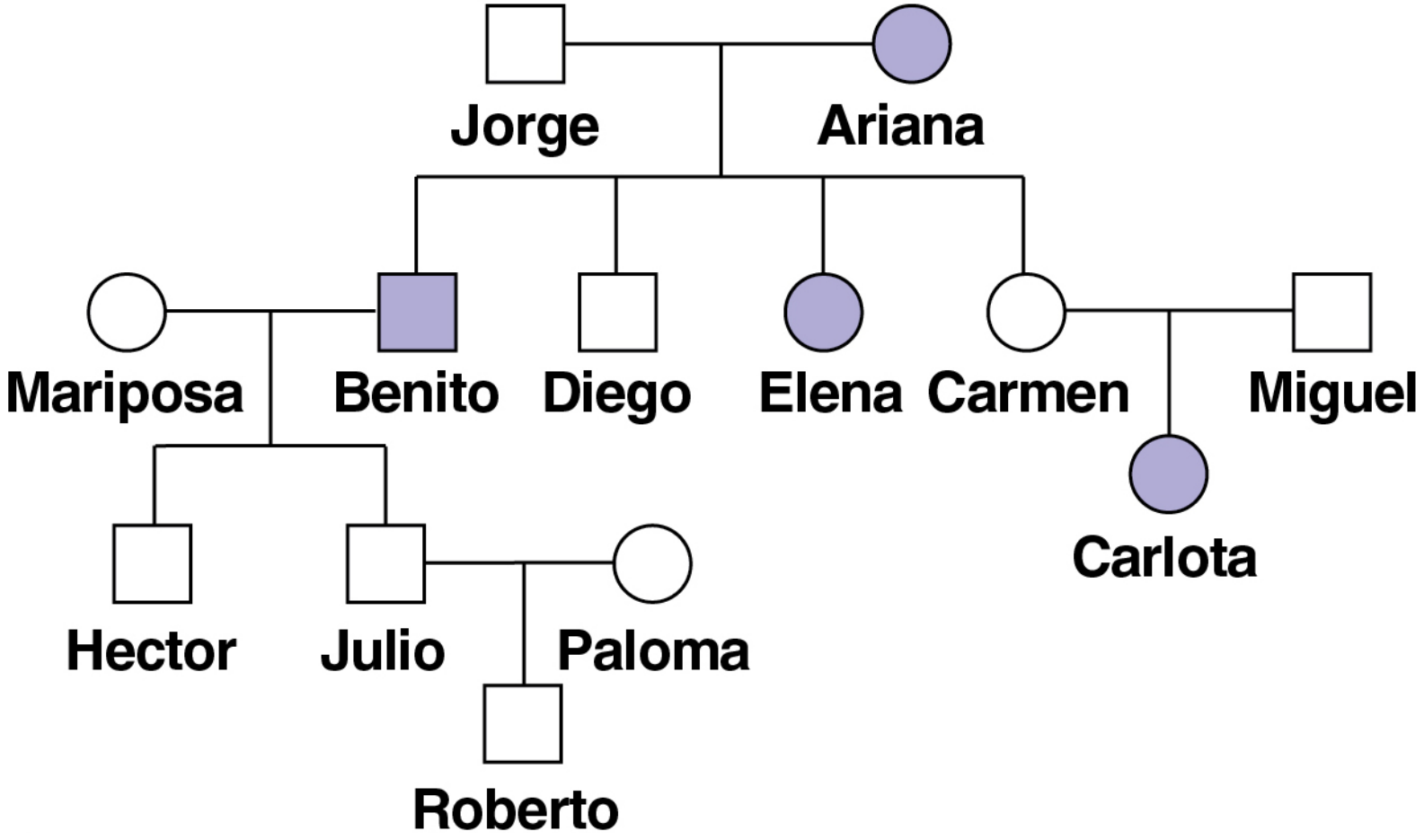
ww
Widow's peak
No widow's peak


Sickle-cell alleles
Low
O_2
Sickle-
cell
disease
Sickle-cell hemoglobin proteins
Part of a fiber of sickle-cell hemo-globin proteins
Long fibers cause red blood cells to be sickle-shaped


Jorge
Ariana
Mariposa
Benito
Diego
Elena
Carmen
Miguel
Carlota
Hector
Julio
Paloma
Roberto