

HUMAN BIOLOGY

Seventeenth Edition

**Sylvia S. Mader
Michael Windelspecht**

Chapter 21 Genetic Inheritance

21.1 Genotype and Phenotype

Learning Outcomes:

- Distinguish between a genotype and a phenotype.
- Define *allele*, *gene*, *dominant*, and *recessive* as they relate to patterns of inheritance.
- Given the genotype of an individual, identify the phenotype.

Genotype ₁

Genotype.

The genes of an individual.

- Genes are segments of DNA that code for a trait.
- Each gene is located in a specific position, or **locus** (*pl.*, loci), on a chromosome.

Allele—an alternate form of a gene.

- That is, if the trait is eye color, one allele is for blue eyes, one is for brown eyes.

Genotype ₂

Genotype, continued.

Alleles are classified as **dominant** or **recessive**.

Dominant alleles mask the expression of recessive alleles.

- If an allele is dominant, only one copy of that allele needs to be present for that trait to be expressed.
- If an allele is recessive, both of the chromosomes must have the recessive allele for it to be expressed.

Genotype ₃

Alleles, continued.

A dominant allele is assigned an uppercase letter; recessive alleles have lowercase letters.

- That is, albinism: the allele for normal pigmentation is A ; for no pigmentation, a .

For each pair of chromosomes, we receive one chromosome from each parent.

- Therefore, we inherit one allele from each parent, resulting in a pair of alleles for each trait.

Genotype ⁴

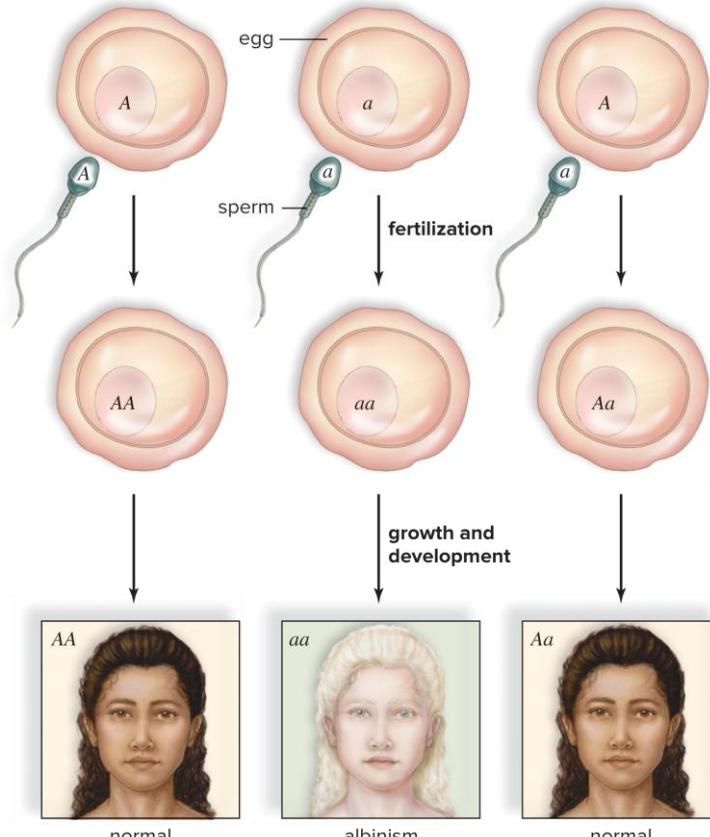
Homozygous dominant—both alleles are dominant (AA).

- The chromosomes of both the sperm and the egg carry the dominant trait.

Homozygous recessive—both alleles are recessive (aa).

Heterozygous—one allele is dominant, one recessive (Aa).

Genetic Inheritance Affects Our Characteristics (Figure 21.1)



Allele Key	
A	Normal pigmentation
a	Lack of pigmentation (albinism)

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Phenotype

Phenotype—the physical appearance of a trait.

- The genotype directs the phenotype.

Check Your Progress 21.1

- Define the following terms: *gene*, *allele*, *locus*, *chromosome*, *dominant*, and *recessive*.
- Describe the difference between genotype and phenotype.
- Summarize the three possible genotypes and their corresponding phenotypes.

21.2 One- and Two-Trait Inheritance ¹

Learning Outcomes:

- Understand how probability is involved in solving one- and two-trait crosses.
- Calculate the probability of a specific genotype or phenotype in an offspring of a genetic cross.

21.2 One- and Two-Trait Inheritance ²

- A **one-trait (monohybrid) cross** examines the patterns of inheritance of only a single set of alleles for a single characteristic.
- A **two-trait cross** explores the patterns of inheritance for two different characteristics.
- For both types of crosses, it is first necessary to determine the gametes of both of the parents in the cross.

Forming the Gametes

When the gametes fuse, the individual has 46 chromosomes (23 homologous pairs; one homologue from each parent).

During meiosis, the homologous chromosomes are separated.

- The gametes have 23 chromosomes.

One-Trait Crosses 1

To predict the chances of having a child with a certain genotype and, therefore, a certain phenotype, consider a **one-trait (monohybrid) cross** involving freckles.

Two parents without freckles have children.

- F —the dominant allele for freckles.
- f —the recessive allele of no freckles.

Examples of Dominant and Recessive Traits (Figure 21.2c,d)



c. Freckles: FF or Ff



d. No freckles: ff

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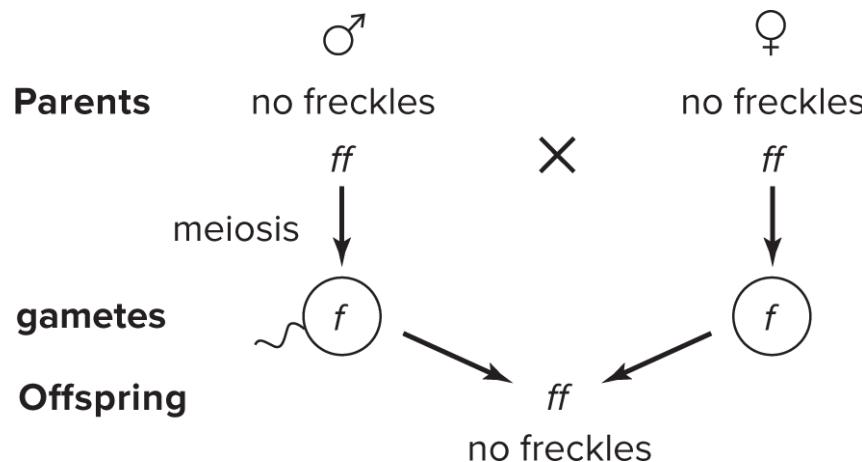
One-Trait Crosses 2

A one-trait cross involving freckles, continued.

If both parents do not have freckles, then their genotypes are both ff .

- The only gametes they can produce contain f .

All the children will be ff and will not have freckles.



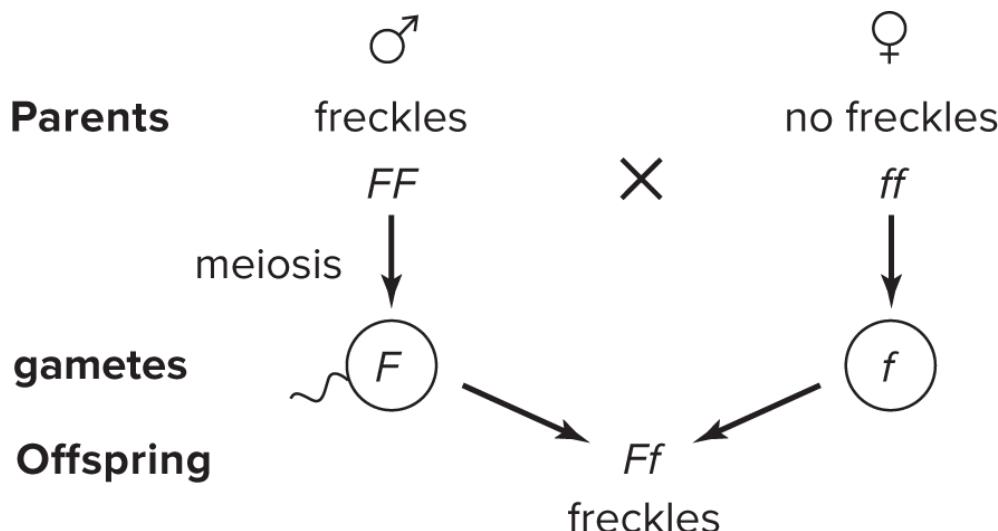
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One-Trait Crosses 3

A one-trait cross involving freckles, concluded.

A homozygous dominant man with freckles (FF) has children with a woman with no freckles (ff).

- The children are heterozygous (Ff) and have freckles.



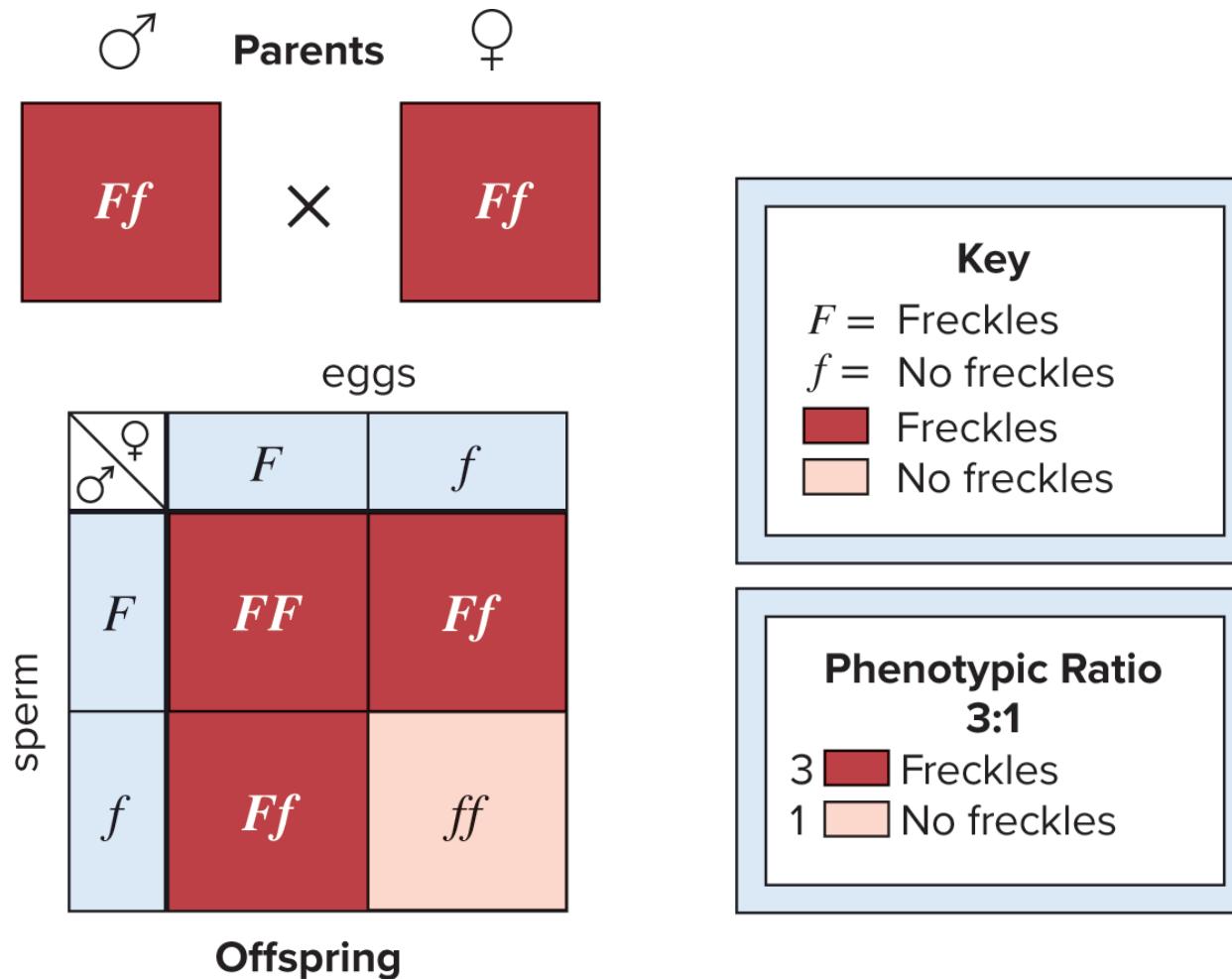
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One-Trait Crosses 4

Punnett square—a tool used to predict the outcome of a cross.

- All possible alleles for the sperm are lined up on one side; the egg alleles are on the other.
- Every possible combination of gametes occurs within the squares.
- In a cross of two heterozygotes ($Ff \times Ff$), each parent has two possible types of gametes (F or f).

Expected Results of a Monohybrid Cross (Figure 21.3)



[Access the text alternative for slide images.](#)

One-Trait Crosses 5

Genotypic and phenotypic ratios.

In the previous slide, the genotypic ratio is 1 FF :2 Ff :1 ff or simply 1:2:1, but the phenotypic ratio is 3:1.

- Prediction: three individuals will have freckles (the FF and the two Ff) and one will not have freckles (the ff).

Each new fertilization is not influenced by any previous fertilizations.

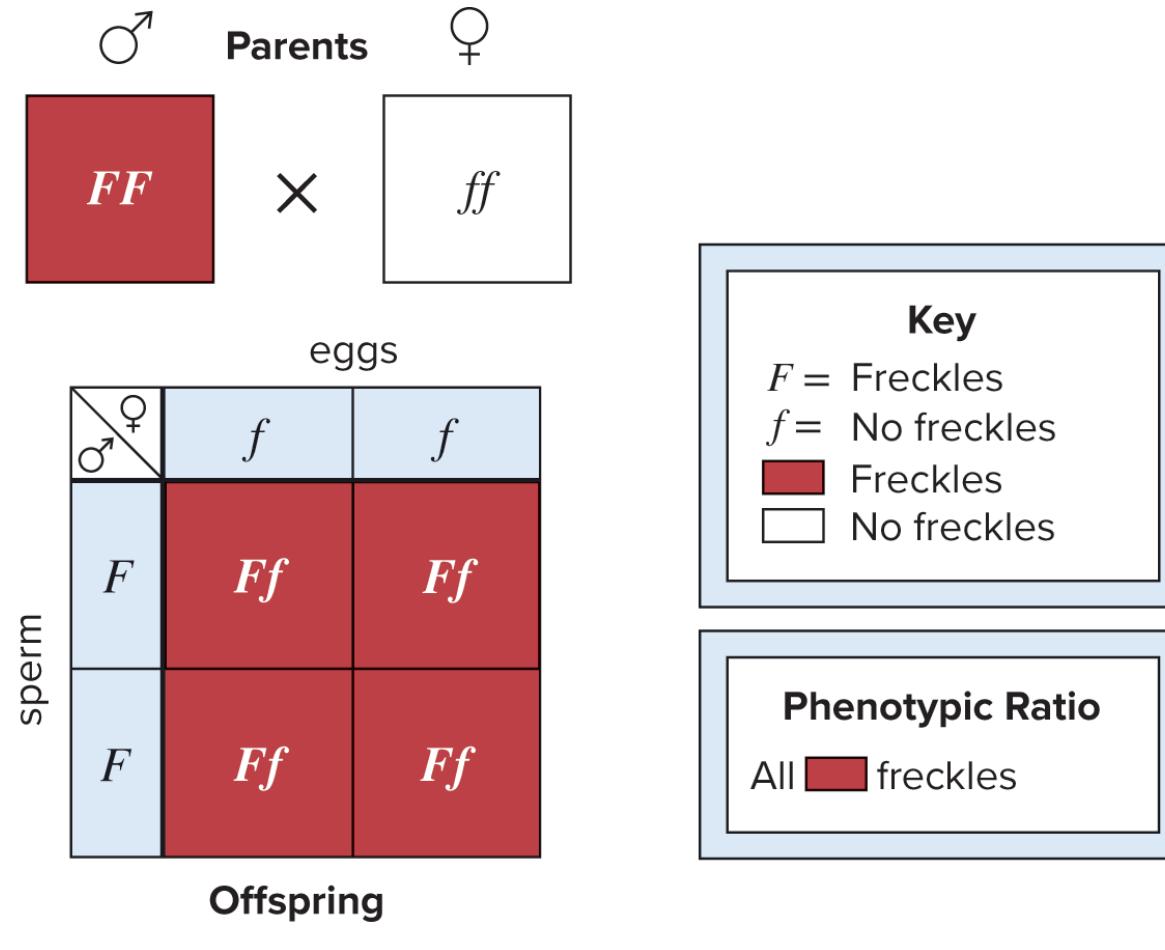
One-Trait Crosses: Determining If the Genotype Is Heterozygous or Homozygous Dominant

A person with a dominant phenotype could be homozygous dominant or heterozygous.

That is, there are two possible results when a man with freckles reproduces with a woman who does not.

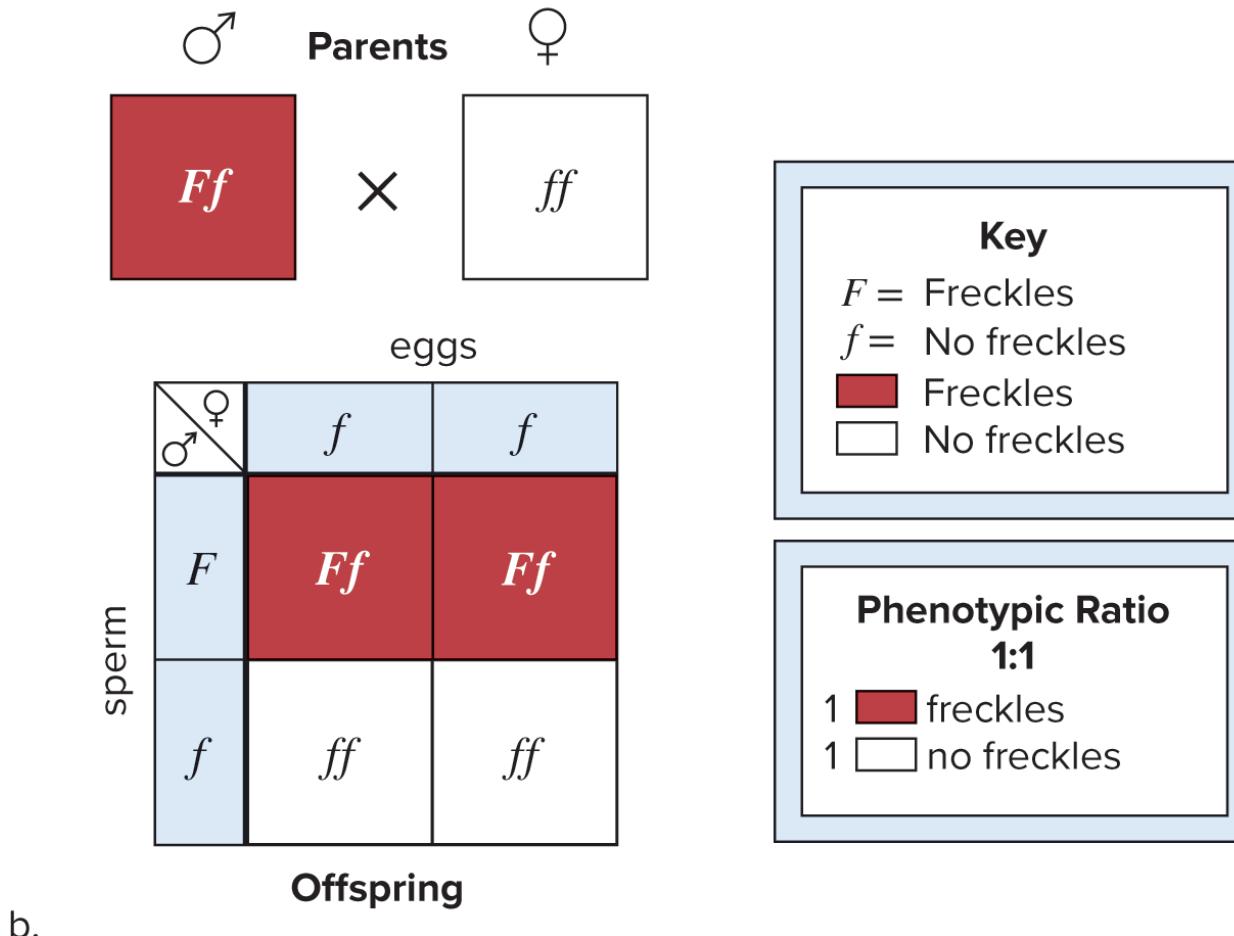
- If the man is FF , all his children will have freckles.
- If the man is Ff , each child has a 50% chance of having freckles.
- The birth of just one child without freckles indicates that the man is heterozygous.

Determining If a Dominant Phenotype Is Homozygous or Heterozygous (Figure 21.4a)



[Access the text alternative for slide images.](#)

Determining If a Dominant Phenotype Is Homozygous or Heterozygous (Figure 21.4b)



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The Punnett Square and Probability ¹

Two laws of probability apply to the study of genetics.

The first is the **product rule**.

- The chance of two different events occurring simultaneously is equal to the multiplied probabilities of each event occurring separately.
- The product rule is often applied to cases in which the word “and” is used.
 - That is, the probability of B *and* C = probability of B multiplied by the probability of C.

The Punnett Square and Probability ²

Two laws of probability apply to the study of genetics, continued.

The second is the **sum rule**.

- Individual probabilities are added to determine total probability for an event.
- The sum rule is often applied to cases in which the word “or” is used.
 - That is, the probability of B *or* C = probability of B plus the probability of C.

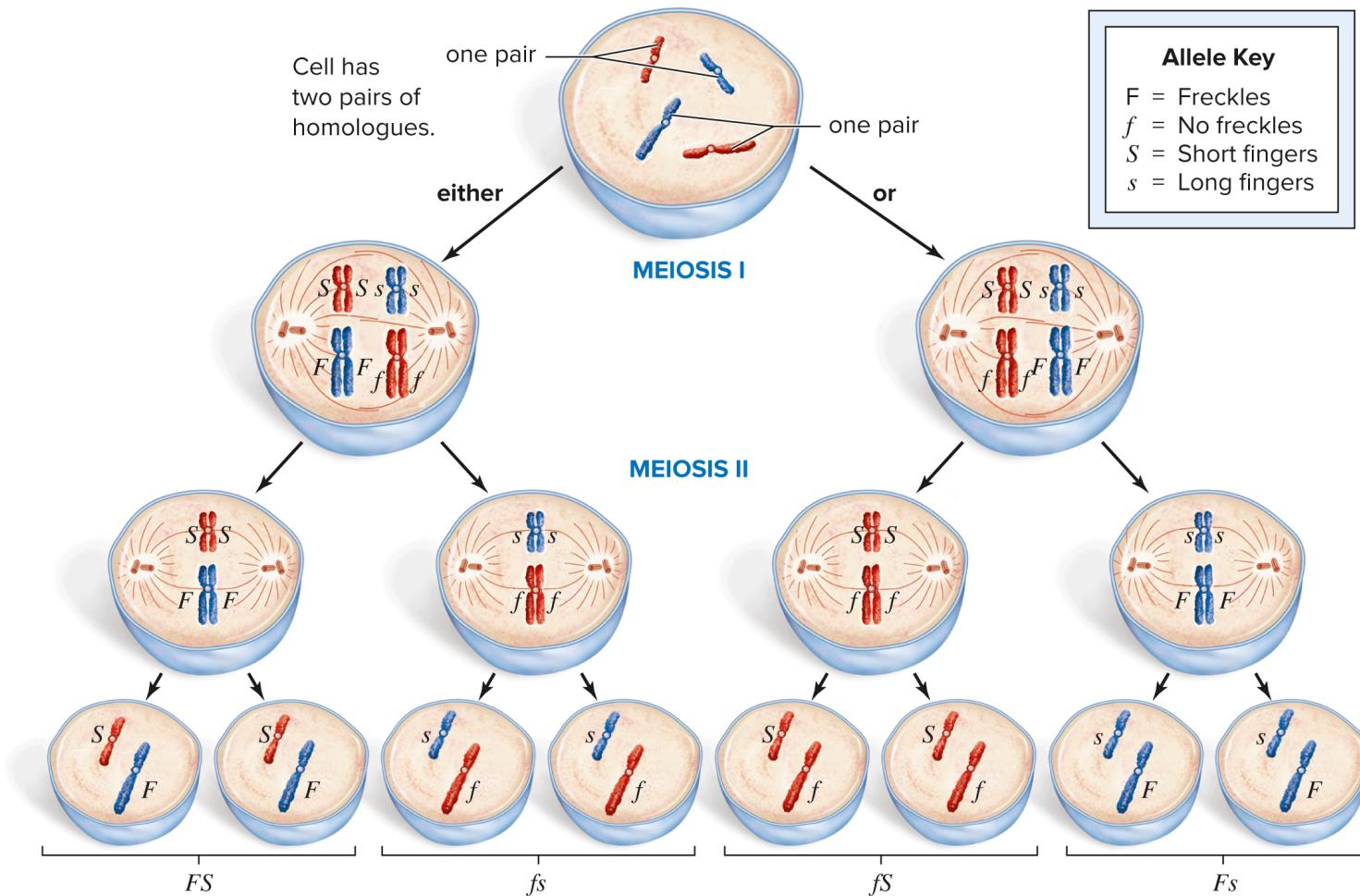
Two-Trait Crosses

Maternal and paternal homologues separate independently during meiosis I.

All possible combinations of alleles occur in the gametes.

- That is, a person with the genotype $FfSs$ would produce the gametes FS , fs , Fs , and fS in equal number.

Meiosis Results in Genetic Diversity of Gametes (Figure 21.5)



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The Dihybrid Cross ₁

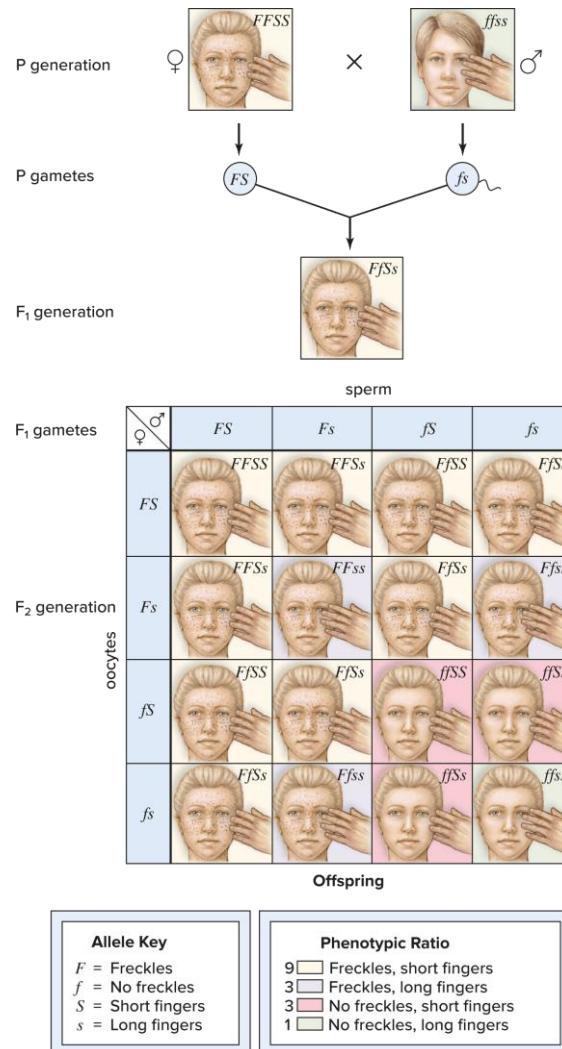
Dihybrid cross.

Involves two traits.

That is, a person homozygous for freckles and short fingers (*FFSS*) reproduces with one who has no freckles and long fingers (*ffss*).

- The gametes for the *FFSS* parent must be *FS* and the gametes for the *ffss* parent must be *fs*.
- Therefore, the offspring will all have the genotype *FfSs* (freckles and short fingers).

Expected Results of a Dihybrid Cross (Figure 21.6)



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The Dihybrid Cross ₂

Dihybrid cross, continued.

When $FfSs$ crosses with $FfSs$, the possible gametes for each is FS , Fs , fS , and fs .

A Punnett square yields:

- 9 freckles and short fingers.
- 3 freckles and long fingers.
- 3 no freckles and short fingers.
- 1 no freckles and long fingers.

This 9:3:3:1 phenotypic ratio is always expected for a dihybrid cross with simple dominance.

Two-Trait Crosses and Probability 1

One can also use the rules of probability to predict the results of a dihybrid cross.

That is, the probable results for two separate monohybrid crosses are as follows:

- For freckles:
 - Probability of freckles = $\frac{3}{4}$.
 - Probability of no freckles = $\frac{1}{4}$.

For finger length:

- Probability of short fingers = $\frac{3}{4}$.
- Probability of long fingers = $\frac{1}{4}$.

Two-Trait Crosses and Probability ²

Using the product rule, we can calculate the probable outcome of a dihybrid cross as follows:

Probability of:

- Freckles and short fingers: $\frac{3}{4} \times \frac{3}{4} = 9/16$.
- Freckles and long fingers: $\frac{3}{4} \times \frac{1}{4} = 3/16$.
- No freckles and short fingers: $\frac{1}{4} \times \frac{3}{4} = 3/16$.
- No freckles and long fingers: $\frac{1}{4} \times \frac{1}{4} = 1/16$.

The expected phenotypic ratio is 9:3:3:1.

Two-Trait Crosses: Determining If the Genotype Is Heterozygous or Homozygous Dominant

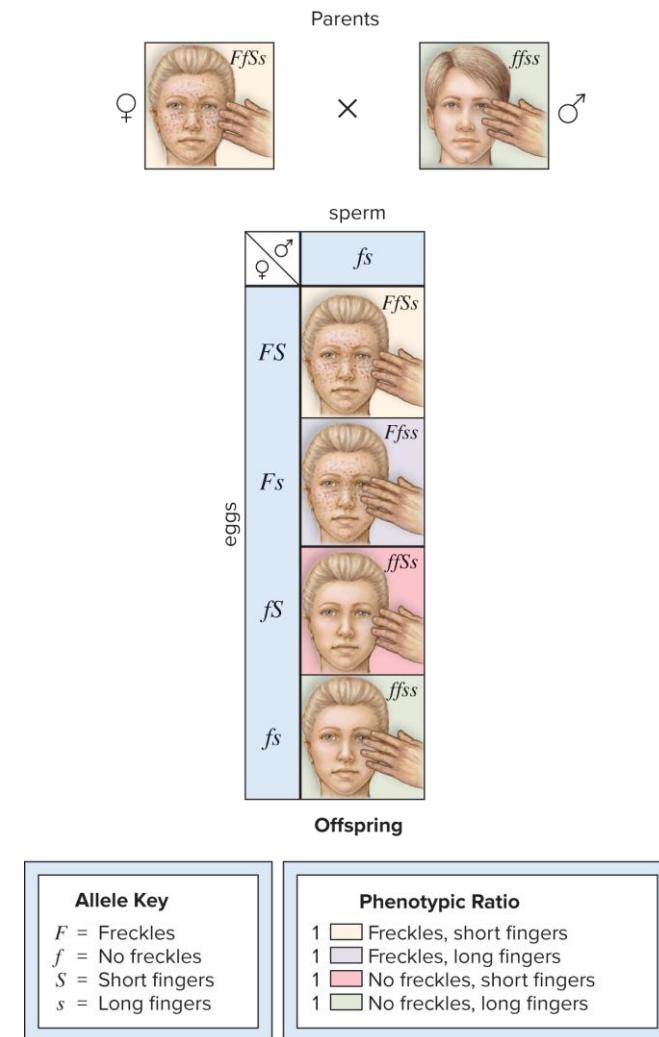
A person with a dominant phenotype for two alleles could be homozygous dominant or heterozygous.

If $FFSS$ reproduces with $ffss$, all the children will have the dominant phenotypes.

If $FfSs$ crosses with $ffss$, each child has a 25% chance of showing one or both recessive traits.

- The expected ratio is 1 freckles with short fingers:1 freckles with long fingers:1 no freckles with short fingers:1 no freckles with long fingers, or 1:1:1:1.

Determining If an Individual Is Homozygous Dominant or Heterozygous (Figure 21.7)



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Phenotypic Ratios of Common Crosses (Table 21.1)

Table 21.1 Phenotypic Ratios of Common Crosses

Genotypes	Phenotypes
Monohybrid $Aa \times$ monohybrid Aa	3:1 dominant to recessive)
Monohybrid $Aa \times$ recessive aa	1.1 (dominant to recessive)
Dihybrid $AaBb \times$ dihybrid $AaBb$	9:3:3:1 (9 both dominant: 3 dominant for one of the traits: 3 dominant for other trait: 1 both recessive)
Dihybrid $AaBb \times$ recessive $aabb$	1:1:1:1 (all possible combinations in equal number)

Check Your Progress 21.2

- Explain how the results of a dihybrid cross are related to the events of meiosis.
- Predict what genotype the children will have if one parent is homozygous recessive for no freckles and homozygous dominant for short finger length ($ffSS$) and the other parent is homozygous dominant for freckles and homozygous recessive for long fingers ($FFss$).
- Using a dihybrid cross as an example (see Fig. 21.6), explain how the gametes are formed by the process of meiosis.

21.3 Inheritance of Genetic Disorders ¹

Learning Outcomes:

- Interpret a human pedigree to identify the pattern of inheritance for a trait.
- Understand the genetic basis of select human autosomal dominant and autosomal recessive genetic disorders.

21.3 Inheritance of Genetic Disorders ²

Many diseases occur as a result of mutations in DNA.

The abnormal gene could be passed down in the sperm or egg.

- The parents may or may not have been affected.

Or the genetic mutation may be new.

21.3 Inheritance of Genetic Disorders ³

Many diseases occur as a result of mutations in DNA, continued.

Autosomal dominant genetic disorder— AA or Aa will have the disorder.

Autosomal recessive genetic disorder—only aa will have the disorder.

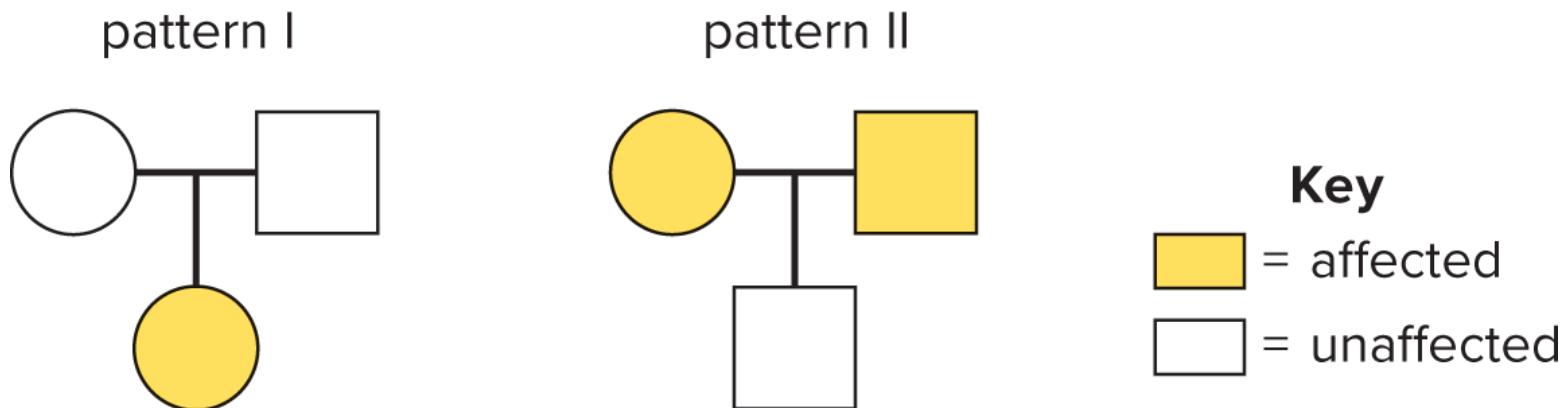
Pedigree—used to determine whether a condition is dominant or recessive.

- Shows the pattern of inheritance.

21.3 Inheritance of Genetic Disorders ⁴

Pedigree.

- Males are squares, females are circles.
- Shaded are affected individuals.
- A line between a square and a circle represents a mating.
- A vertical line going downward leads to offspring.



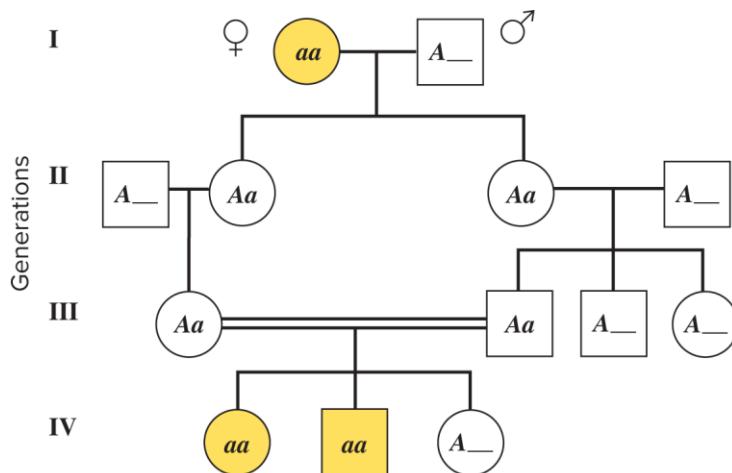
Autosomal Recessive Patterns of Inheritance 1

Carrier—a heterozygote; carries the recessive trait but the phenotype is dominant.

With autosomal recessive traits, if both parents are affected, all the children are affected.

- The parents can pass on only recessive alleles.
- All children will also be homozygous recessive.

Autosomal Recessive Disorder Pedigree (Figure 21.8)



Key	
aa	affected
Aa	carrier (unaffected)
AA	unaffected
$A-$	unaffected (one allele unknown)

Autosomal recessive disorders

- Most children who are affected have parents who are not affected.
- Heterozygotes (Aa) have an unaffected phenotype.
- Two parents who are affected will always have children who are affected.
- Close relatives who reproduce are more likely to have children who are affected.

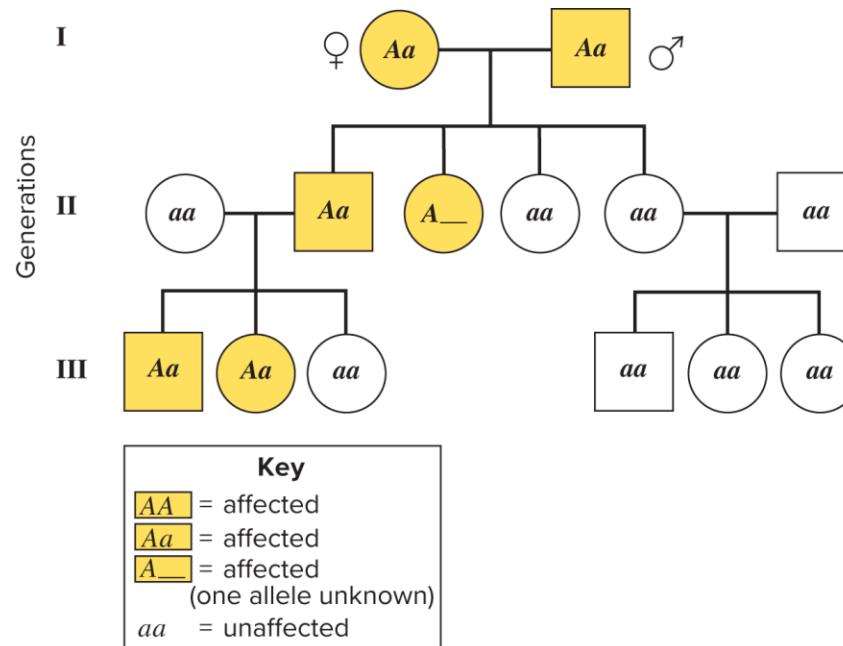
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Autosomal Recessive Patterns of Inheritance 2

If a condition is dominant, the parents can be heterozygous and still be affected.

- A child can inherit a recessive allele from each parent and be unaffected.
- When a disorder is dominant, an affected child must have at least one affected parent.

Autosomal Dominant Disorder Pedigree (Figure 21.9)



Autosomal dominant disorders

- Children who are affected will usually have a parent who is affected.
- Heterozygotes (Aa) are affected.
- Two parents who are affected can produce a child who is not affected.
- Two parents who are unaffected will not have children who are affected.
- Both males and females are affected with equal frequency.

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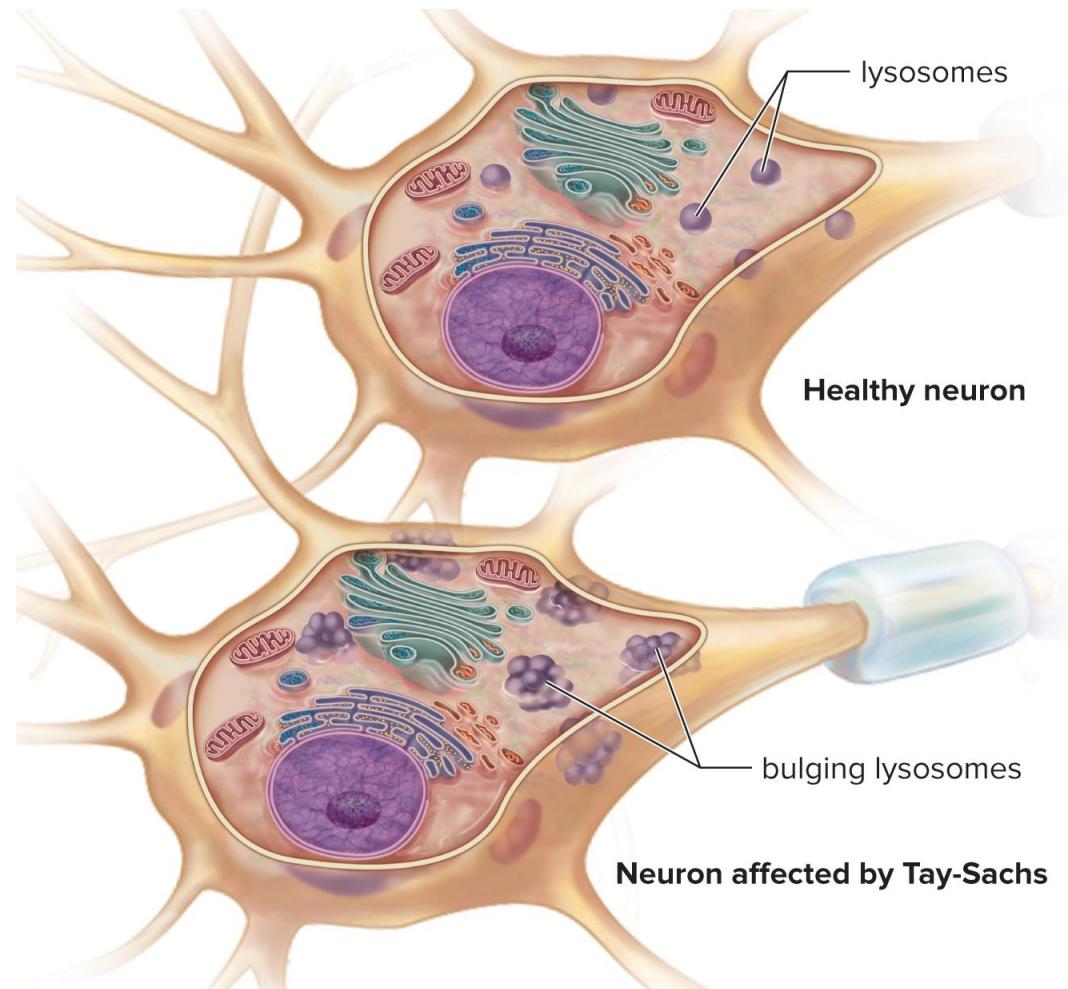
Autosomal Recessive Disorders ¹

With an **autosomal recessive disorder**, affected individuals are *aa*.

That is, **Tay-Sachs disease**.

- Occurs among Ashkenazic Jewish people and their descendants.
- Results from a lack of a lysosome enzyme, hex A, which clears out fatty acid proteins in the brain.
- Causes blindness, seizures, and paralysis.
- There is no cure; children affected die by age 5.

Neuron Affected by Tay-Sachs Disease (Figure 21.10)



Autosomal Recessive Disorders ₂

That is, **cystic fibrosis (CF)**.

Most prevalent in whites.

Caused by a defective chloride channel that is encoded by the *cystic fibrosis conductance transmembrane regulator (CFTR)* allele on chromosome 7.

- 1 in 29 whites in the United States carries this allele.

Chloride ions (Cl^-) can't pass through the defective CFTR chloride channel.

Autosomal Recessive Disorders ³

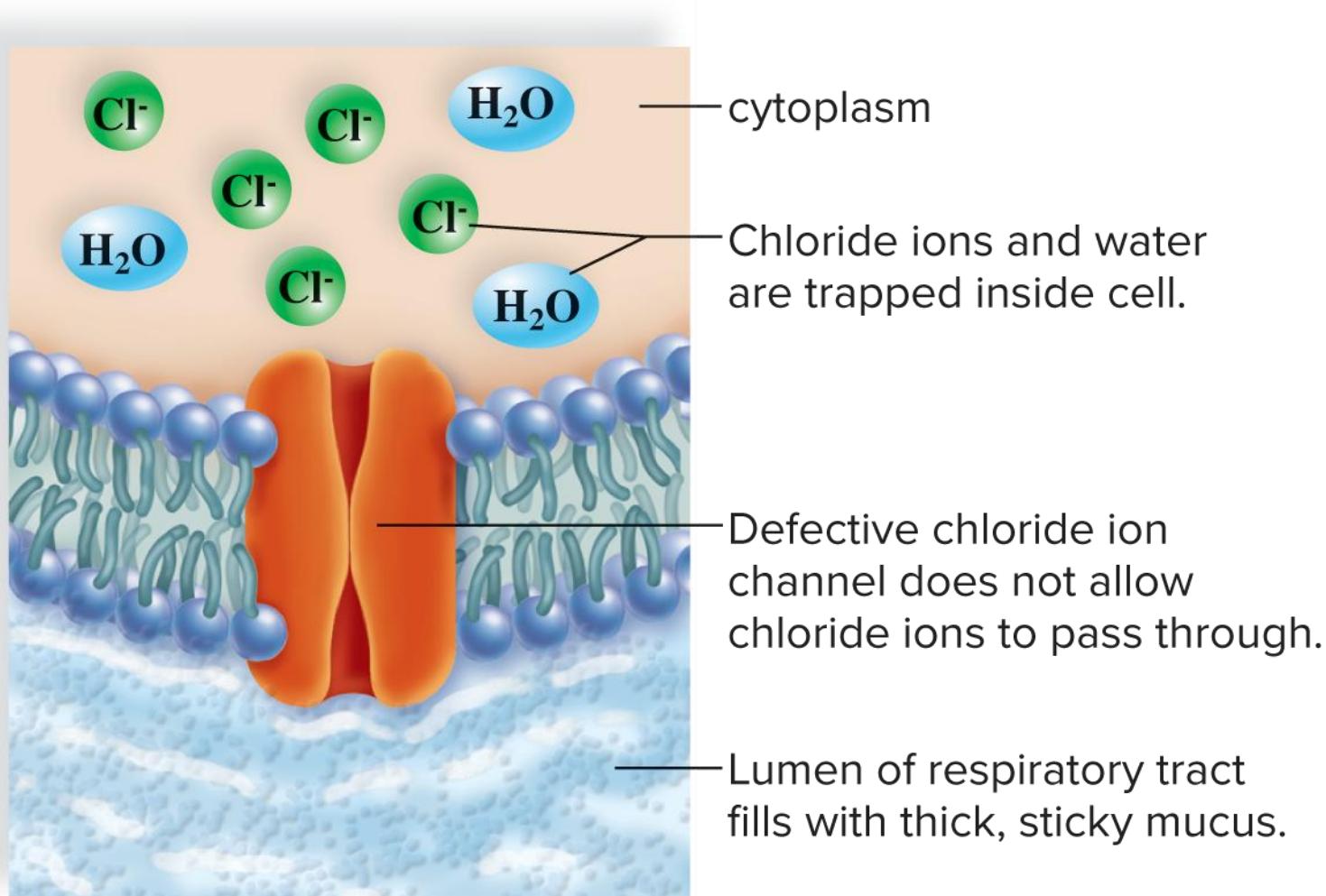
Cystic fibrosis (CF), continued.

Ordinarily, after Cl^- passes through the channel to the other side of the membrane, Na^+ and water follow.

- If Cl^- can't pass through, water can't either.
- Causes abnormally thick mucus to form in the bronchial tubes and pancreatic ducts.

Gene therapy has been successful in treating some forms of CF.

Cystic Fibrosis Disease (Figure 21.11)



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Autosomal Recessive Disorders ⁴

That is, **sickle-cell disease**.

- Autosomal recessive disorder.
- Red blood cells are sickle-shaped.
- They live for only 2 weeks (should live 4 months).
- Caused by abnormal hemoglobin that differs by only one amino acid.
- Prevalent among African Americans.
- Sickle-shaped cells can't pass through narrow capillaries, so they clog the vessels, preventing adequate circulation.

Autosomal Recessive Disorders ⁵

That is, **sickle-cell disease**, continued.

Results in anemia, tissue damage, jaundice, joint pain, and gallstones.

Treatment: blood transfusions, bone marrow transplants.

Heterozygotes—RBCs are normal unless they experience dehydration or oxygen deprivation.

- Experience episodes and symptoms like homozygous patients.

Autosomal Dominant Disorders ₁

In an **autosomal dominant disorder**, affected individuals have only one abnormal allele.

That is, **Marfan syndrome**.

- Caused by a defect in the production of an elastic connective tissue protein called **fibrillin**.
- Symptoms: a dislocated lens, long limbs and fingers, a caved-in chest, weakened wall of the aorta.
- Treatments: beta-blockers to control the cardiovascular symptoms, corrective lenses or eye surgery, and braces or orthopedic surgery for musculoskeletal symptoms.

Autosomal Dominant Disorders ²

That is, **osteogenesis imperfecta**.

Weakened, brittle bones.

Mutations in two genes for type I collagen—one of the most abundant proteins in the human body.

- Collagen provides strength and rigidity to bone and forms the framework for most tissues.

Autosomal Dominant Disorders 3

That is, **Huntington's disease**.

Progressive degeneration of brain cells.

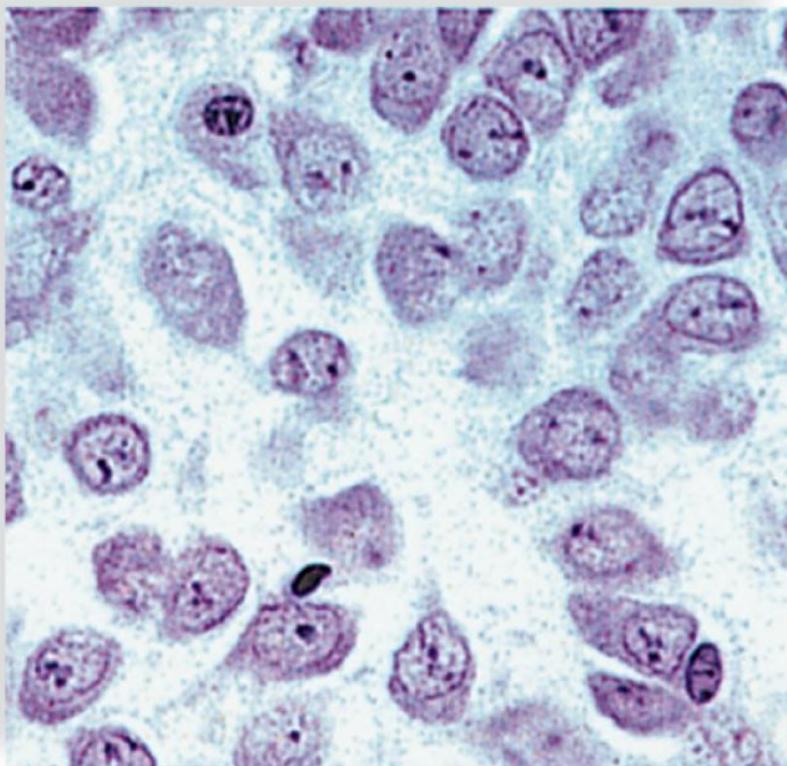
Caused by a mutation of the protein **huntingtin**.

- Huntington forms clumps inside neurons that render them inactive.

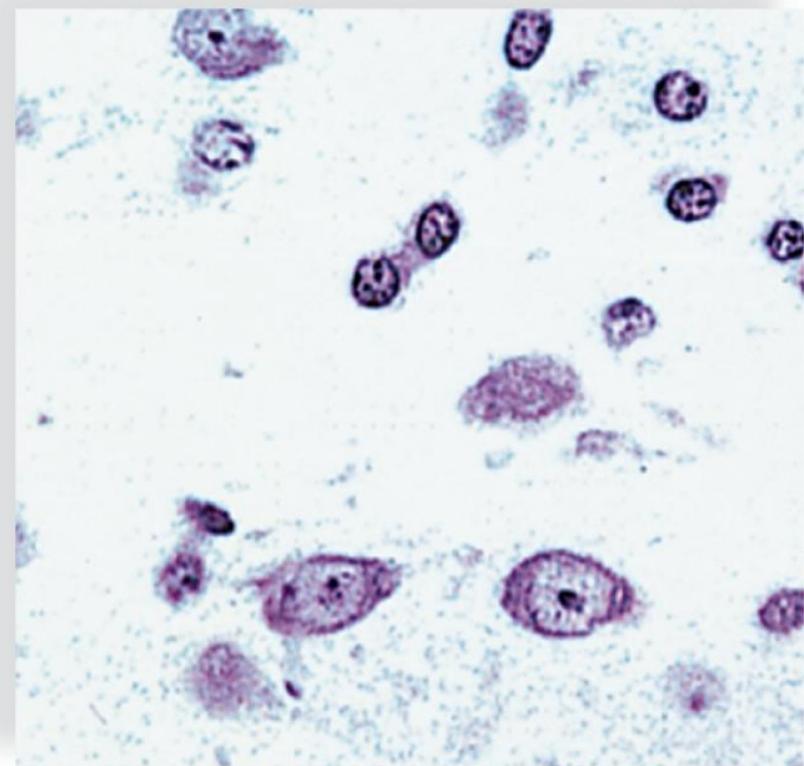
Symptoms don't appear until the late 30s to late 40s.

- Uncontrolled movements, unsteady gait, dementia, and speech impairment.
- Patients live 15 to 20 years after onset of symptoms.

Huntington's Disease (Figure 21.12)



many neurons in normal brain



loss of neurons in Huntington's brain

Check Your Progress 21.3

- Solve the following: In a pedigree, all the members of one family are affected. Based on this knowledge, list the genotypes of the parents (a) if the trait is recessive and (b) if the trait is dominant.
- Predict the chances that homozygous normal parents for cystic fibrosis will have a child with cystic fibrosis.
- Explain why some incidences of autosomal recessive disorders are higher in one race or culture.

21.4 Beyond Simple Inheritance Patterns

Learning Outcomes:

- Summarize how polygenic inheritance, pleiotropy, codominance, and incomplete dominance differ from simple one-trait crosses.
- Explain how a combination of genetics and the environment can influence a phenotype.
- Predict a person's blood type based on the genotype.

Polygenic Inheritance

Polygenic traits—controlled by several sets of alleles.

That is, skin color and height.

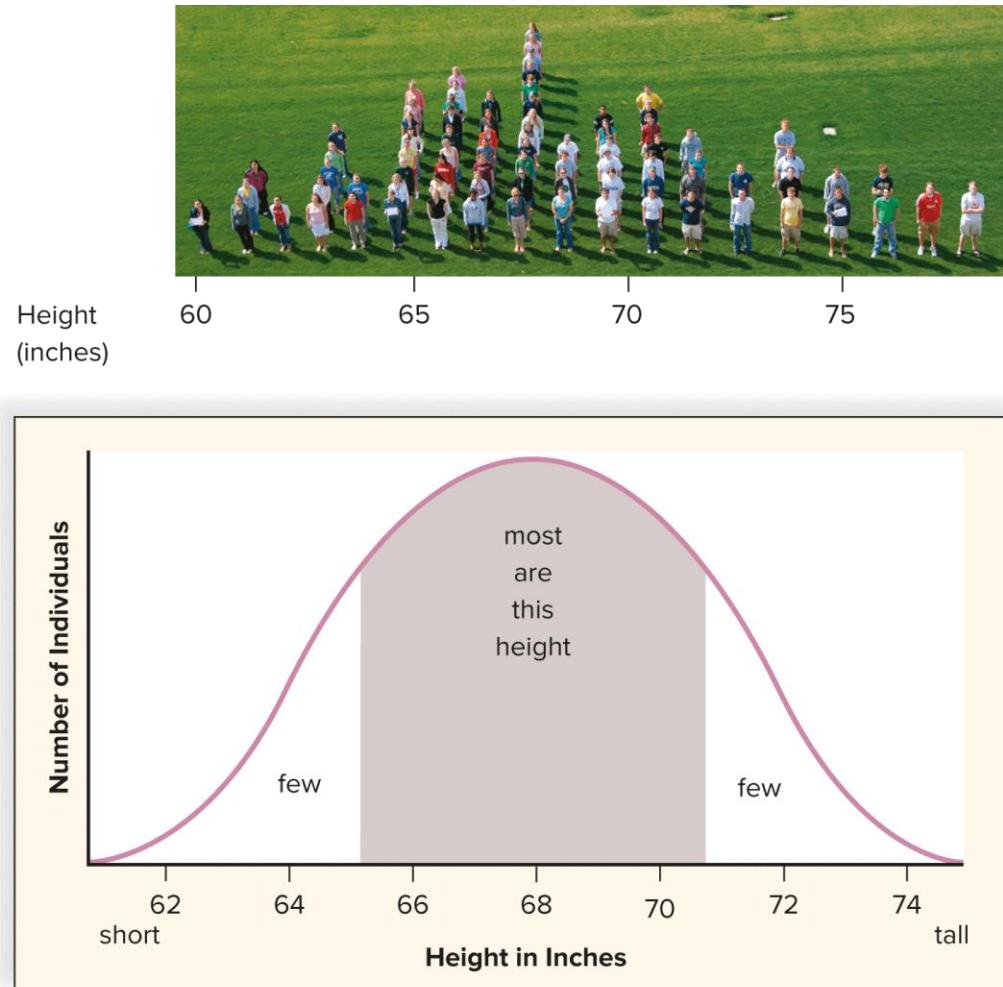
The effects of each dominant allele are additive.

- **Continuous variation** of phenotypes, resulting in a distribution that resembles a bell-shaped curve.

Environmental effects cause many intervening phenotypes.

- That is, nutrition influences height.

Height Is a Polygenic Trait in Humans (Figure 21.13)



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Skin Color

Skin color.

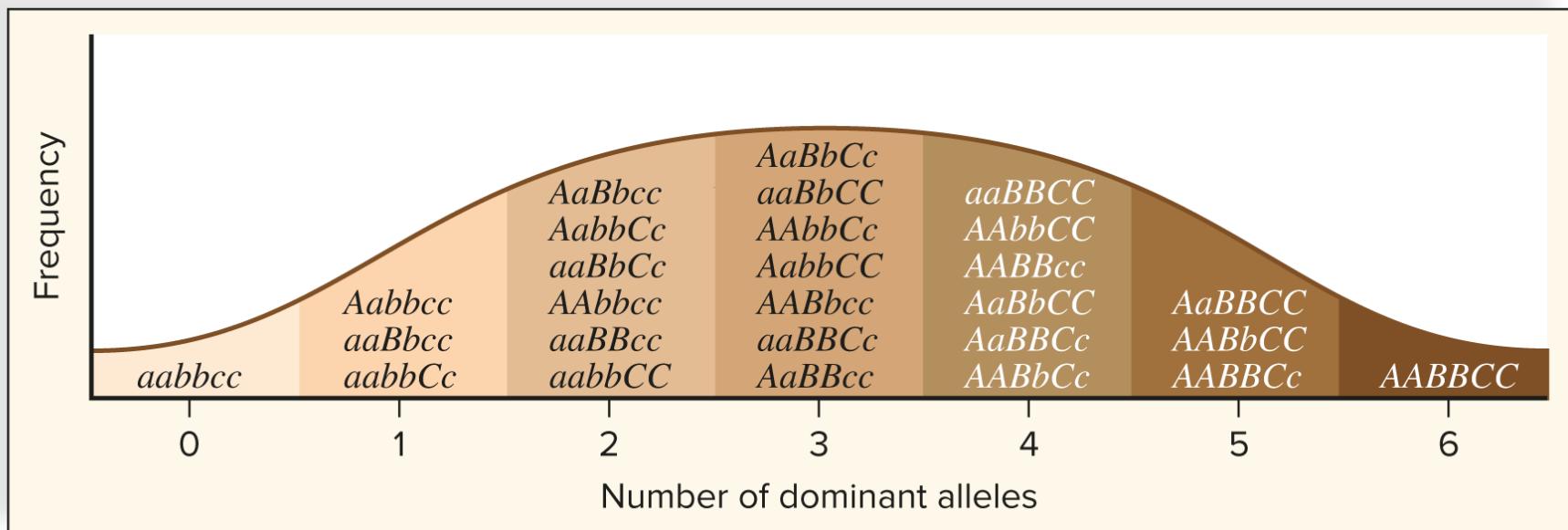
The result of pigmentation produced by melanocytes.

There are over 100 different genes that influence skin color.

Is influenced by the sunlight in the environment.

- A range of phenotypes exists for each genotype.

Polygenic Inheritance and Skin Color (Figure 21.14)



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Environmental Influence

Multifactorial traits—controlled by polygenes subject to environmental influences.

That is, cleft lip and/or palate, clubfoot, schizophrenia, diabetes, allergies, and cancers.

That is, the coats of Himalayan rabbits are darker at the ears, nose, paws, and tail.

- The enzyme that makes melanin is active only at low temperatures.

Himalayan Rabbit with Temperature-Susceptible Coat Color (Figure 21.15)

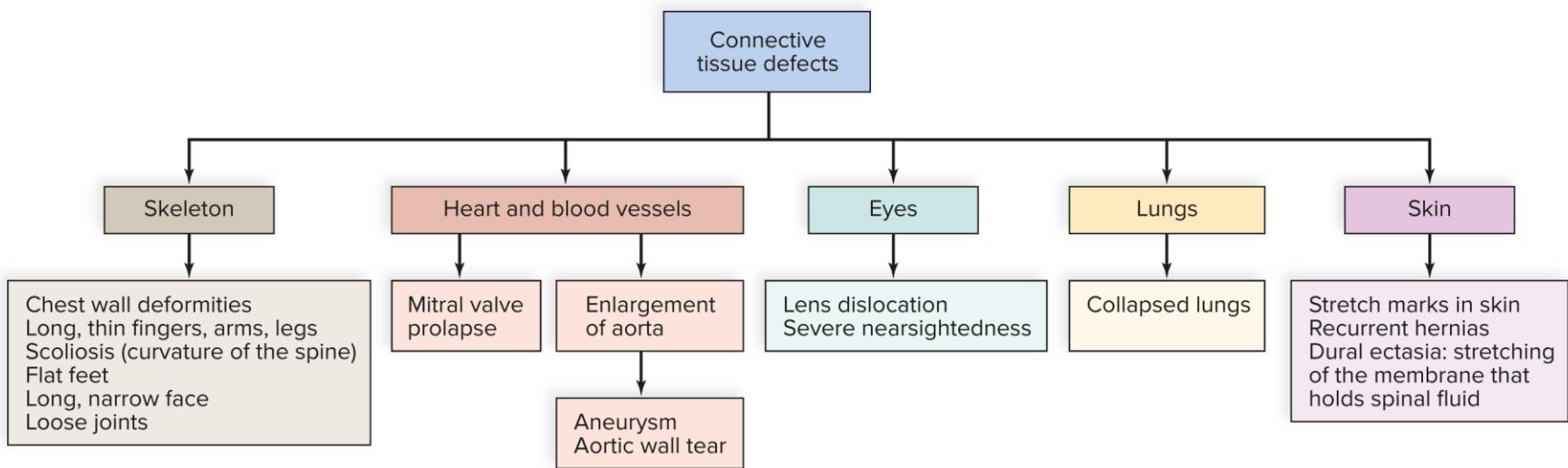


Pleiotropy

Pleiotropy—a single mutant gene affects two or more distinct and seemingly unrelated traits.

- That is, Marfan syndrome has widespread effects all over the body.

Marfan Syndrome (Figure 21.16)



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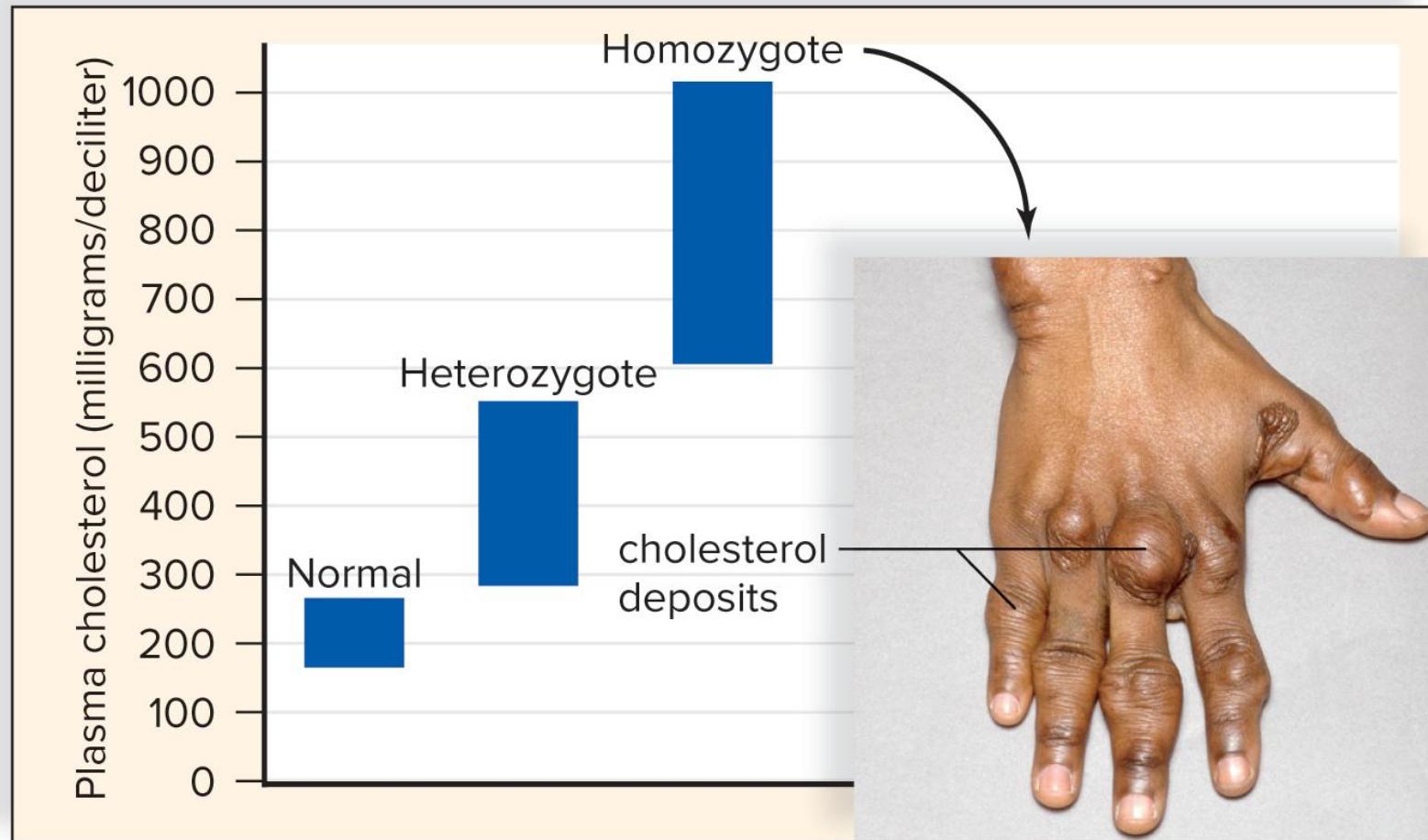
Incomplete Dominance and Codominance 1

Incomplete dominance example:

Familial hypercholesterolemia—associated with a gene that controls the number of LDL cholesterol receptor proteins in parallels in the plasma membrane of cells.

- A person with two mutated alleles lacks LDL-cholesterol receptors.
 - Often die of cardiovascular disease before age 30.
 - Develop cholesterol deposits in the skin, tendons, and cornea.
- A person with only one mutated allele has half the normal number of receptors.
 - May die while still young or after they have reached middle age.
- A person with two normal alleles has the usual number of receptors.
 - Do not have familial hypercholesterolemia.

The Inheritance of Familial Hypercholesterolemia (Figure 21.17)



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Incomplete Dominance and Codominance ²

Codominance—alleles are equally expressed in a heterozygote.

- That is, blood type AB: RBCs have the characteristics of both type A and type B blood.

Multiple-Allele Inheritance 1

When a trait is controlled by **multiple alleles**, the gene exists in several allelic forms.

Each person has only two of the possible alleles.

That is, **ABO blood types**.

- Each person has only two of the three possible alleles, and both I^A and I^B are dominant over i .
- There are two possible genotypes for type A blood and two possible genotypes for type B blood.

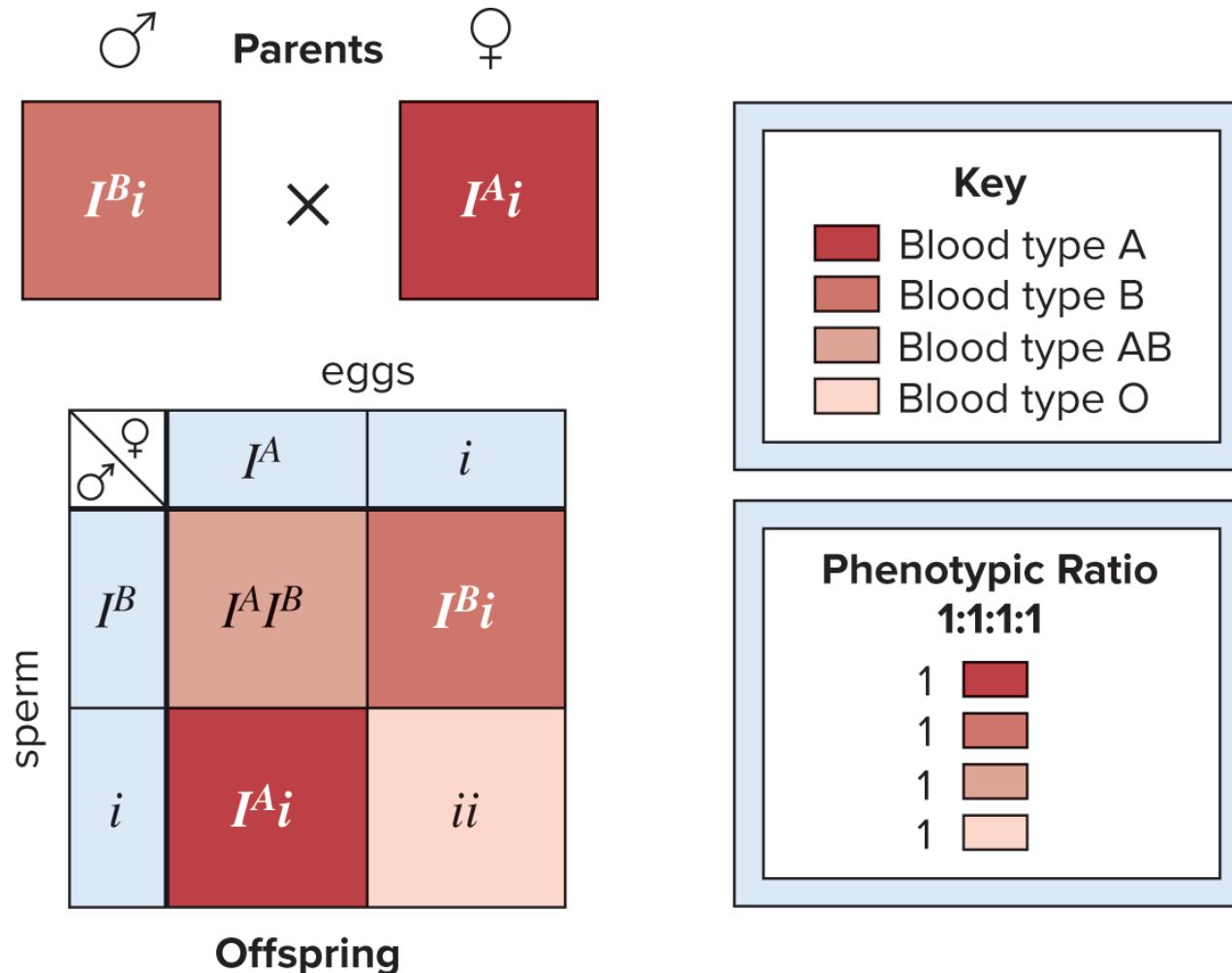
Multiple-Allele Inheritance 2

ABO blood types, continued.

- I^A and I^B are fully expressed when together.
- Therefore, if a person inherits one of each of these alleles, that person will have type AB blood.
- Type O blood results from two i alleles.

Phenotype	Possible Genotypes
A	$I^A I^A, I^A i$
B	$I^B I^B, I^B i$
AB	$I^A I^B$
O	ii

The Inheritance of ABO Blood Types (Figure 21.18)



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Check Your Progress 21.4

- Detail why polygenic inheritance follows a bell-shaped curve.
- Describe a multifactorial trait that could have diet and nutrition as environmental influences.
- Discuss the potential evolutionary advantages of having multiple alleles for a trait.

21.5 Sex-Linked Inheritance 1

Learning Outcomes:

- Understand the differences between autosomal and sex-linked patterns of inheritance.
- Interpret a human pedigree to determine the sex-linked inheritance of a trait.

21.5 Sex-Linked Inheritance 2

Both males and females have 23 pairs of chromosomes; 22 pairs are called **autosomes**, and the other pair are the **sex chromosomes**.

Sex chromosomes differ between the sexes: males have X and Y, and females have two X chromosomes.

- The Y chromosome contains the gene responsible for determining male gender.

21.5 Sex-Linked Inheritance 3

Sex-linked traits—controlled by genes on the sex chromosomes.

An allele on an X chromosome is **X-linked**.

An allele on the Y chromosome is **Y-linked**.

- Most sex-linked genes are on the X chromosome.

The X carries genes that affect both males and females.

21.5 Sex-Linked Inheritance 4

Sex-linked traits, continued.

A male always receives an X-linked allele from the mother, from whom the X chromosome is inherited.

- The Y chromosome from the father does not carry the same genes as an X chromosome.

Usually, the alleles associated with sex-linked genetic disorders are recessive.

- Therefore, a female must receive two alleles, one from each parent, to have the disorder.

X-Linked Alleles

For X-linked traits, the allele is shown as a letter attached to the X chromosome.

Genotypes	Phenotypes
$X^B X^B$	Female who has normal color vision
$X^B X^b$	Carrier female who has normal color vision
$X^b X^b$	Female who is color blind
$X^B Y$	Male who has normal color vision
$X^b Y$	Male who is color blind

X-Linked Alleles: Color Blindness ₁

Although a female carrier has normal color vision, she can pass on the allele for color blindness.

- To be color blind, females must receive the allele from both parents.
- Color blindness in males is more common; they need only one recessive allele to be color blind.

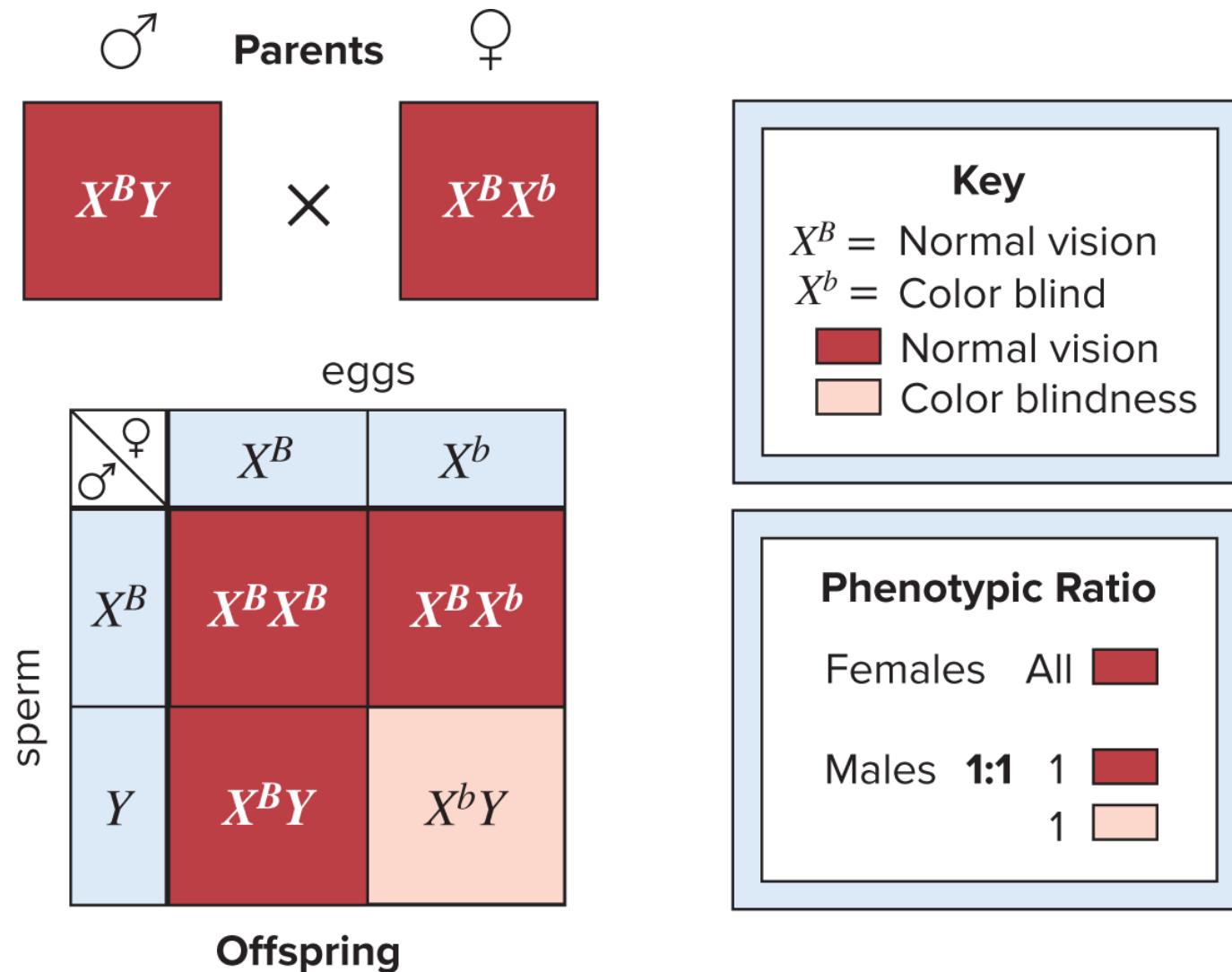
X-Linked Alleles: Color Blindness 2

The allele for color blindness must be inherited from the mother, because it is on the X chromosome.

- Males only inherit the Y chromosome from the father.

The inheritance of a Y from the father cannot offset the inheritance of an X^b from the mother.

Results of an X-Linked Cross (Figure 21.19)



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Pedigree for X-Linked Disorders ¹

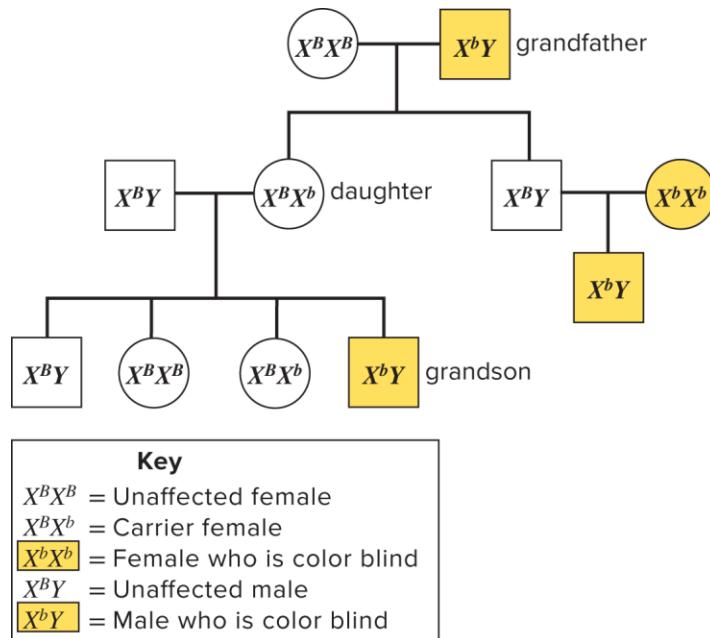
X-linked recessive disorder.

More males than females have it, because recessive alleles on the X chromosome are always expressed in males.

- The Y chromosome lacks an allele for the disorder.

X-linked recessive conditions often pass from grandfather to grandson, because the daughters of a male with the disorder are carriers.

X-Linked Recessive Disorder Pedigree (Figure 21.20)



X-linked recessive disorders

- More males than females are affected.
- A son who is affected can have parents who have the normal phenotype.
- For a female to have the characteristic, her father must also have it. Her mother must have it or be a carrier.
- The characteristic often skips a generation from the grandfather to the grandson.
- If a woman has the characteristic, all of her sons will have it.

[Access the text alternative for slide images.](#)

Pedigree for X-Linked Disorders ²

X-linked dominant traits.

Males with the condition pass the trait *only* to daughters, who have a 100% chance of having the condition.

- Females can pass an X-linked dominant allele to both sons and daughters.

X-Linked Recessive Disorders of Interest ₁

Color blindness.

The inability to differentiate between certain colors.

- Some forms (red-green) are X-linked.
- There is no treatment.
- Affects about 1 in 12 males and 1 in 200 females in the United States.

X-Linked Recessive Disorders of Interest ²

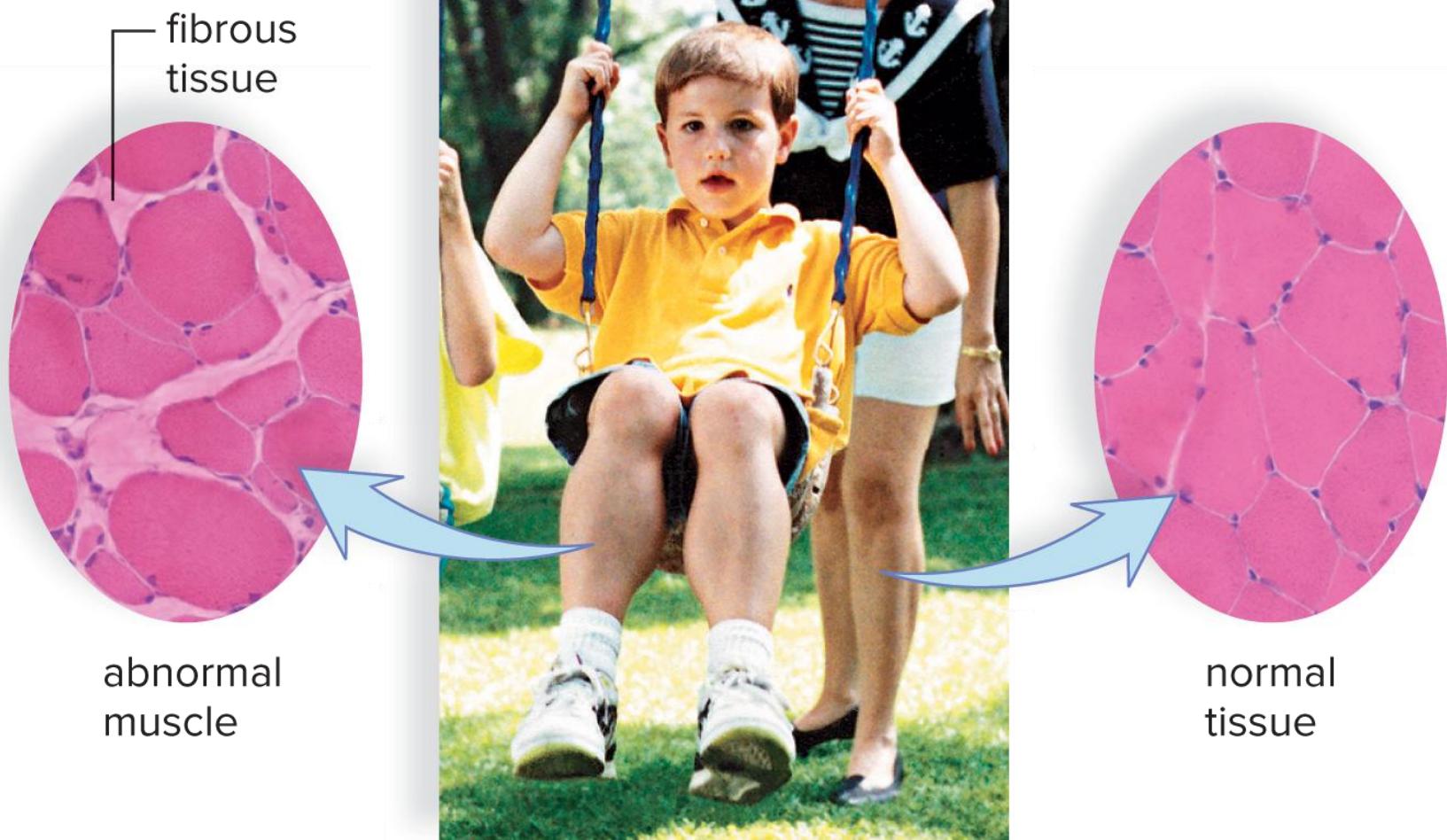
Duchenne muscular dystrophy.

X-linked recessive.

Degeneration of the muscles: waddling gait, toe walking, frequent falls, and difficulty in rising.

- Progresses until wheelchair use is required, normally by ages 7–10.
- Death usually occurs by ages 20–25.
- Therefore, affected males are rarely fathers; the allele remains in the population through passage from carrier mother to carrier daughter.

Muscular Dystrophy (Figure 21.21)



X-Linked Recessive Disorders of Interest ³

Duchenne muscular dystrophy, continued.

Cause: the absence of a protein called **dystrophin**.

- Calcium leaks into the cell, which stimulates an enzyme that dissolves muscle fibers.

X-Linked Recessive Disorders of Interest ⁴

Fragile X syndrome.

X-linked recessive.

- Most common cause of inherited intellectual disability.
- Impairment varies in severity.
- The most common known cause of **autism spectral disorder**.
- Physical characteristics: prominent jaw, flexible joints, and genital abnormalities.
- Also a dislike of being touched, poor eye contact, repetitive speech patterns, and hand flapping.

X-Linked Recessive Disorders of Interest ⁵

Hemophilia.

X-linked recessive disorder.

- Two common types: **hemophilia A** and **B**.
 - A—absence of clotting factor VIII.
 - B—absence of clotting factor IX.
- Blood does not clot well.
- Hemorrhages can be stopped with transfusions of fresh blood (or plasma) or the clotting protein.

Check Your Progress 21.5

- Solve the following: In a given family, a man and woman have two children, a boy and a girl, and both are color-blind. List the possible genotypes of the parents if both parents have normal vision.
- Can a woman who is affected by an X-linked dominant disorder have a child who is not affected? Why or why not?
- Discuss why X-linked disorders are more common than Y-linked disorders.