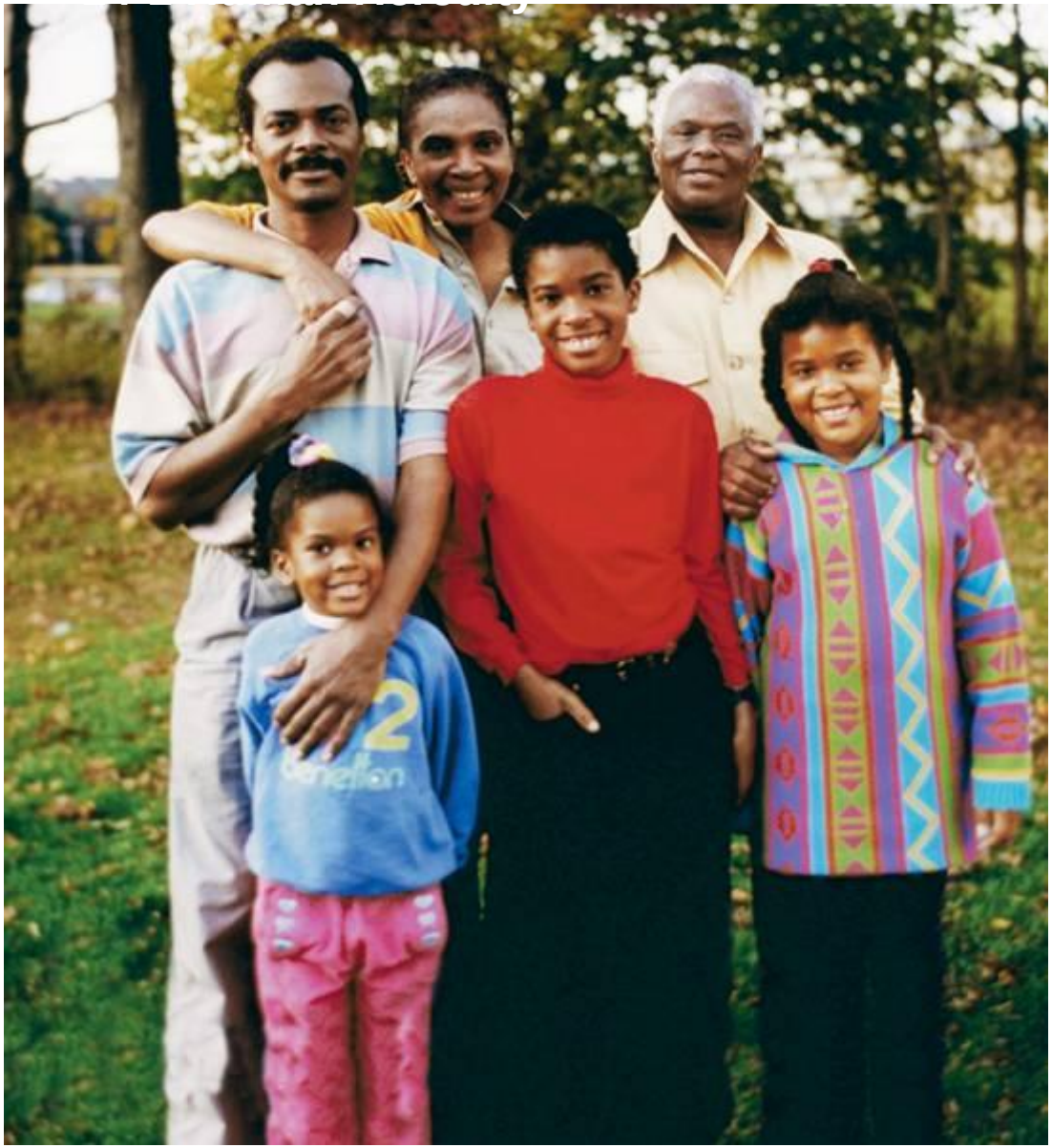
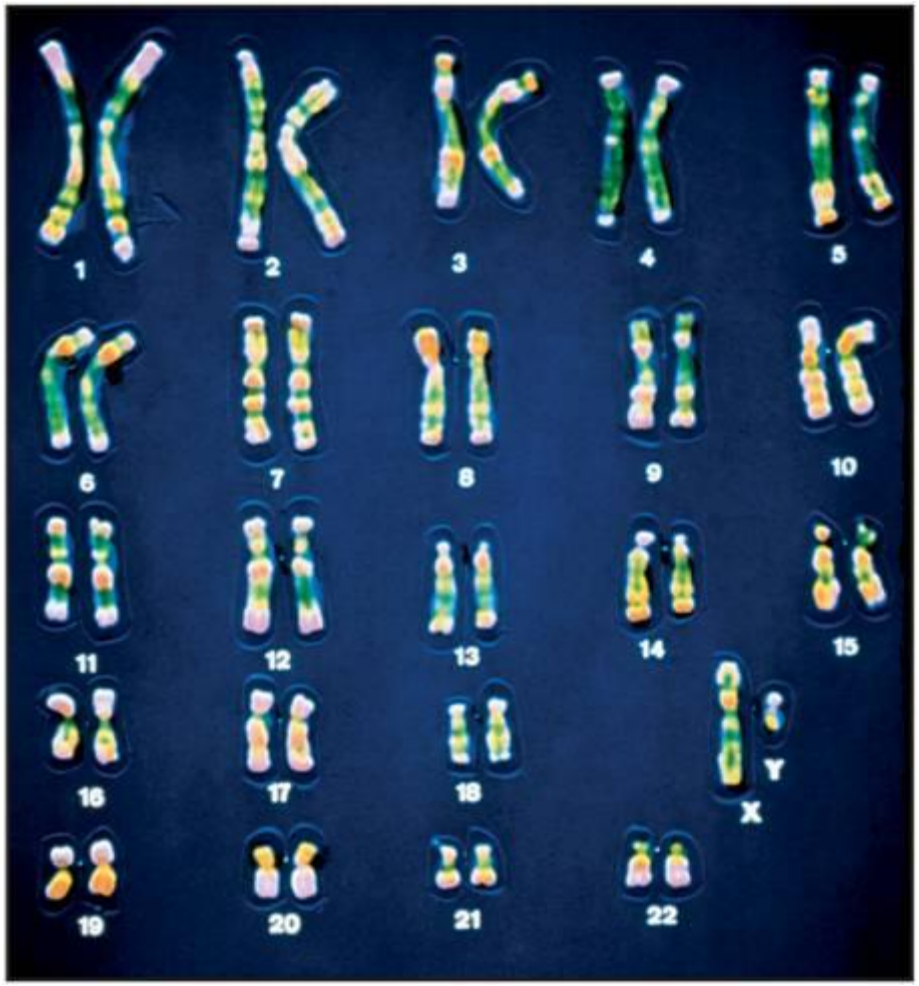
14–1 Human Heredity



Human Karyotype



- Females have two copies of an X chromosome.
- Males have one X chromosome and one Y chromosome.

The remaining 44 chromosomes are known as **autosomes**.

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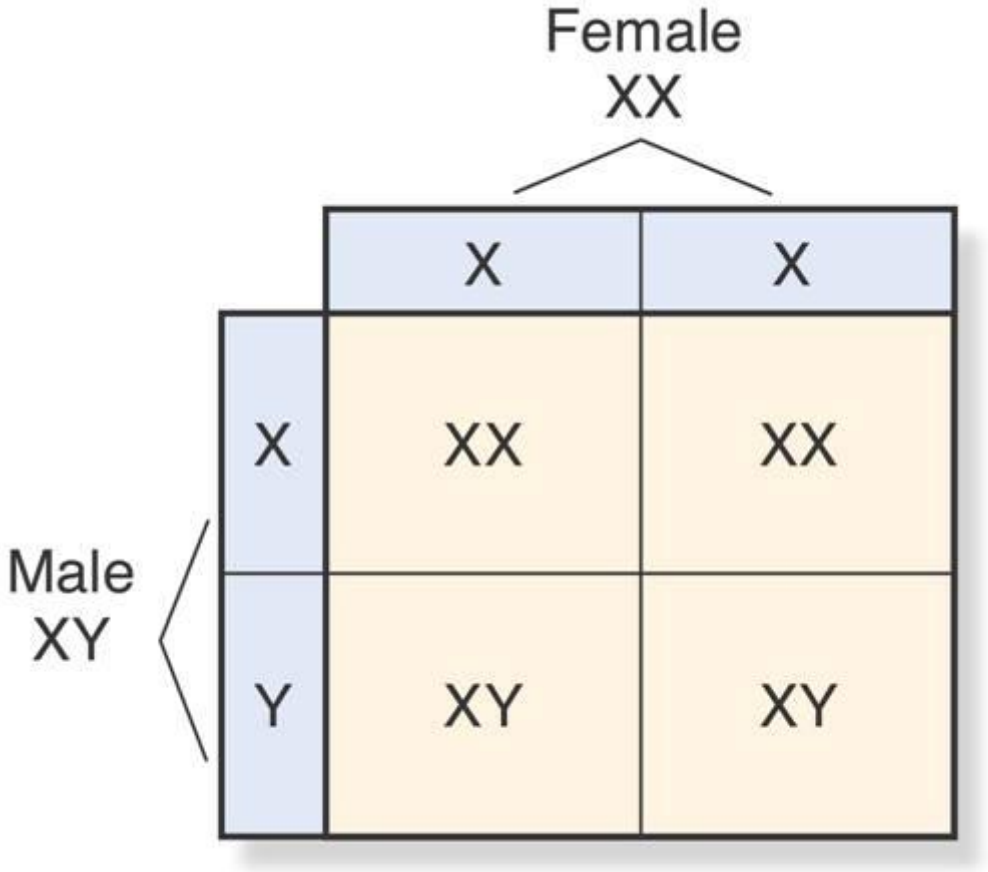
All human egg cells carry a single X chromosome (23,X).

Half of all sperm cells carry an X chromosome (23,X) and half carry a Y chromosome (23,Y).

About half of the zygotes will be 46,XX (female) and half will be 46,XY (male).



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Pedigree Charts

A **pedigree** chart shows the relationships within a family.

Genetic counselors analyze pedigree charts to infer the genotypes of family members.

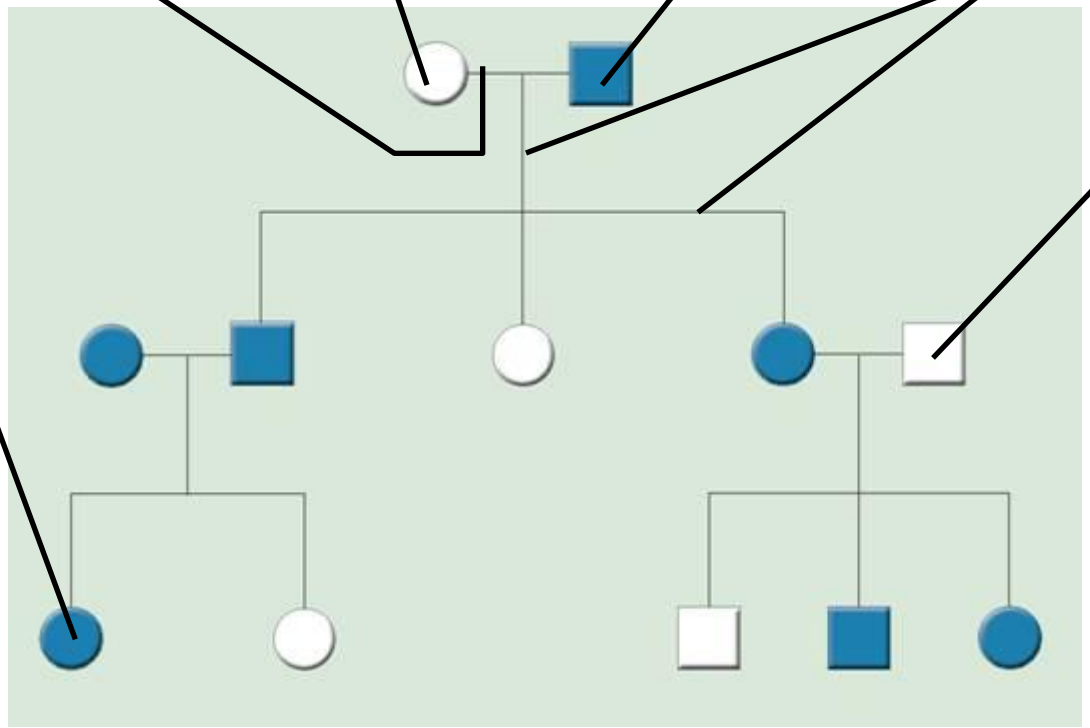
A horizontal line connecting a male and a female indicates that a person is a carrier for the trait.

A circle represents a female.

A square represents a male.

A vertical line and a bracket connect the parents to their children.

A shaded circle or square indicates that a person expresses the trait.



A circle or square that is not shaded indicates that a person does not express the trait.

Blood Group Genes

→a variety of genetically determined blood groups.

→The best known are the ABO blood groups and the Rh blood groups.

The Rh blood group is determined by a single gene with two alleles—positive and negative.

The positive (Rh^+) allele is dominant, so individuals who are Rh^+/Rh^+ or Rh^+/Rh^- are said to be

Rh positive.

Individuals with two Rh^- alleles are said to be

Rh negative.

ABO blood group

- There are three alleles for this gene, I^A, I^B, and i.
- Alleles I^A and I^B are codominant.

Individuals with alleles I^A and I^B produce both A and B antigens, making them blood type AB.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
AB	I^AI^B	A and B	AB	A, B, AB, O

The *i* allele is recessive.

Individuals with alleles I^AI^A or I^Ai produce only the A antigen, making them blood type A.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
A	I^AI^A or I^Ai	A	A, AB	A, O

Individuals with I^BI^B or I^Bi alleles are type B.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
B	I^BI^B or I^Bi	B	B, AB	B, O

Individuals who are homozygous for the *i* allele (*ii*) produce no antigen and are said to have blood type O.

Blood Groups

Phenotype (Blood Type)	Genotype	Antigen on Red Blood Cell	Safe Transfusions	
			To	From
O	*ii*	none	A, B, AB, O	O

Disorders Caused by Recessive Alleles

Disorder	Major Symptoms
Albinism	Lack of pigment in skin, hair, and eyes
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections
Galactosemia	Accumulation of galactose (a sugar) in tissues; mental retardation; eye and liver damage
Phenylketonuria (PKU)	Accumulation of phenylalanine in tissues; lack of normal skin pigment; mental retardation
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in early childhood

Disorders Caused by Dominant Alleles

Disorder	Major Symptoms
Achondroplasia	Dwarfism (one form)
Huntington disease	Mental deterioration and uncontrollable movements; symptoms usually appear in middle age
Hypercholesterolemia	Excess cholesterol in blood; heart disease

Disorders Caused by Codominant Alleles

Disorder	Major Symptoms
Sickle cell disease	Misshapen, or sickled, red blood cells; damage to many tissues

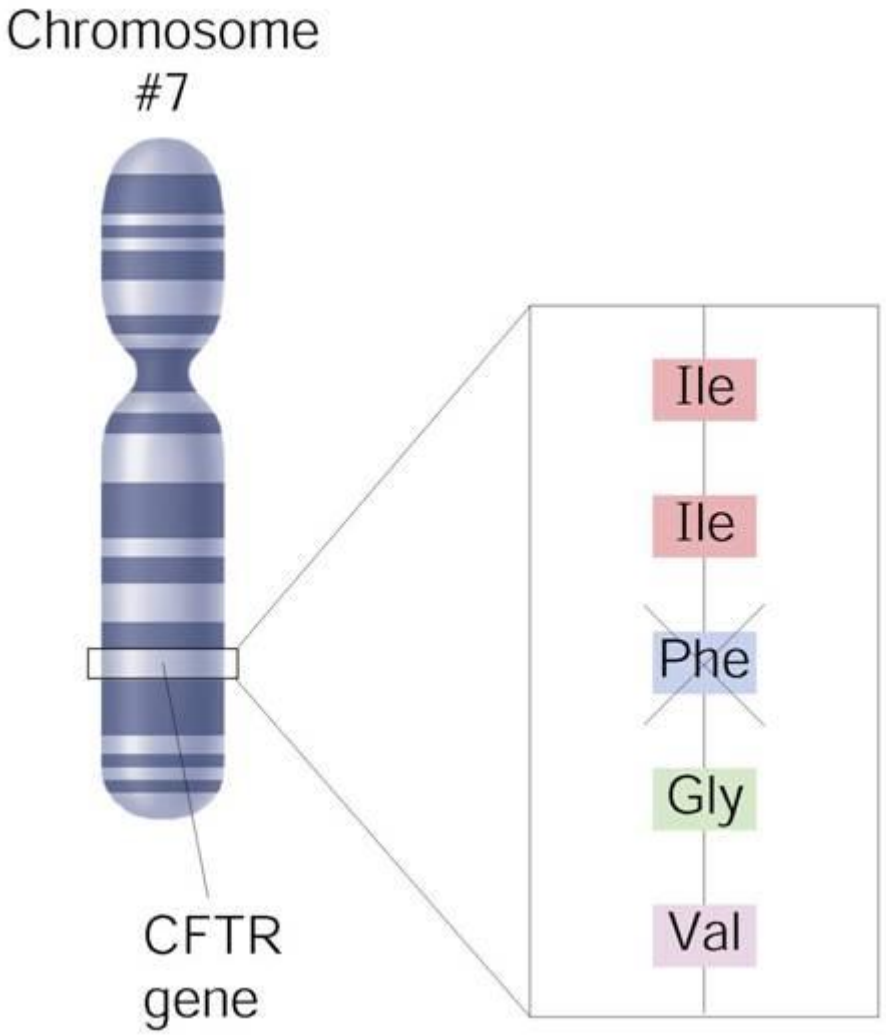
From Gene to Molecule

How do small changes in DNA cause genetic disorders?

In both cystic fibrosis and sickle cell disease, a small change in the DNA of a single gene affects the structure of a protein, causing a serious genetic disorder.

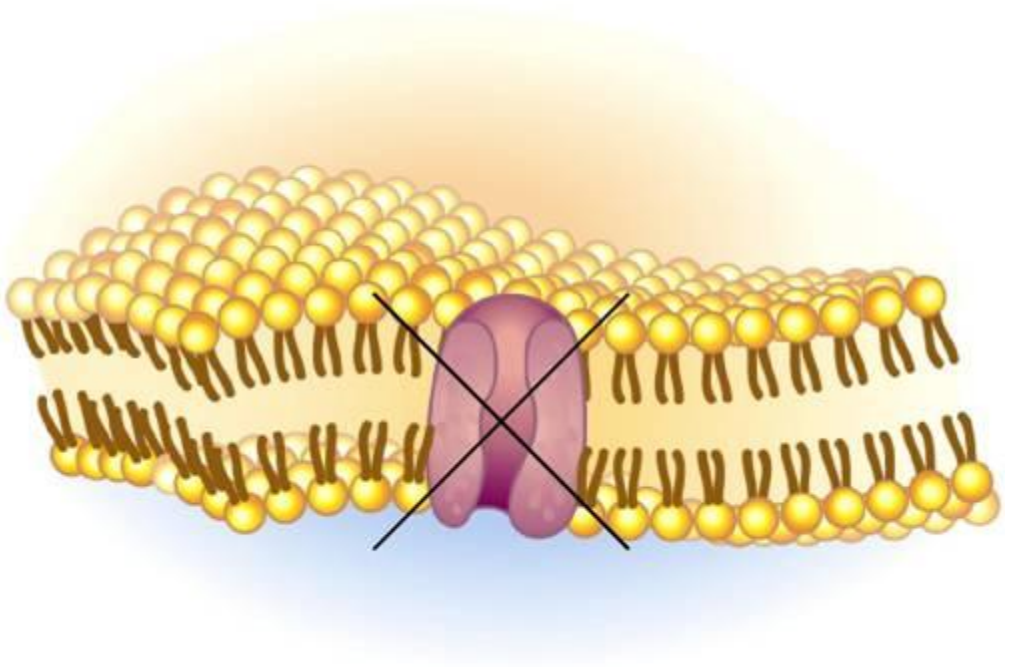
The most common allele that causes cystic fibrosis is missing 3 DNA bases.

As a result, the amino acid phenylalanine is missing from the CFTR protein.



Normal CFTR is a chloride ion channel in cell membranes.

Abnormal CFTR cannot be transported to the cell membrane.



Sickle Cell Disease

Sickle cell disease is a common genetic disorder found in African Americans.

It is characterized by the bent and twisted shape of the red blood cells.

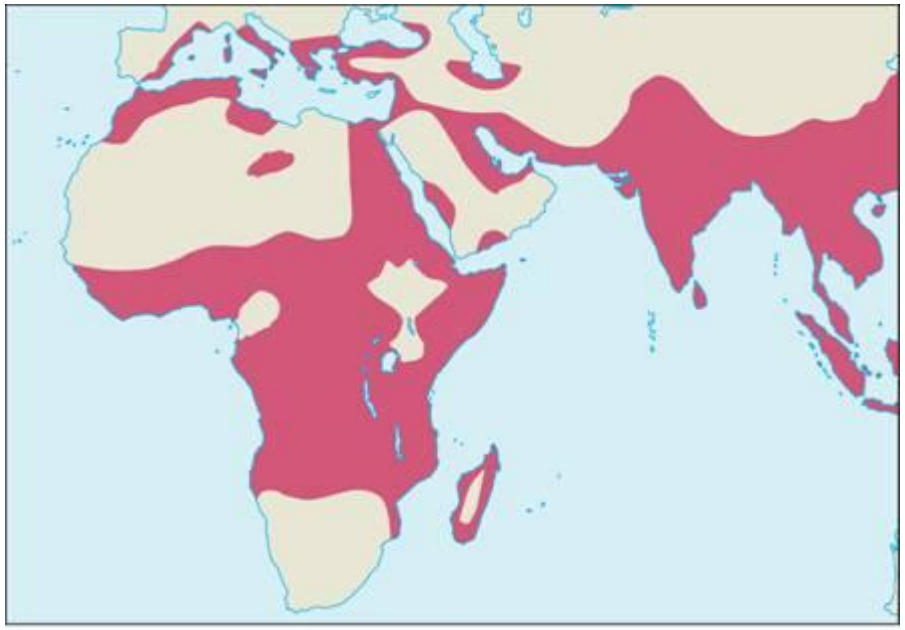
In the sickle cell allele, just one DNA base is changed.

As a result, the abnormal hemoglobin is less soluble than normal hemoglobin.

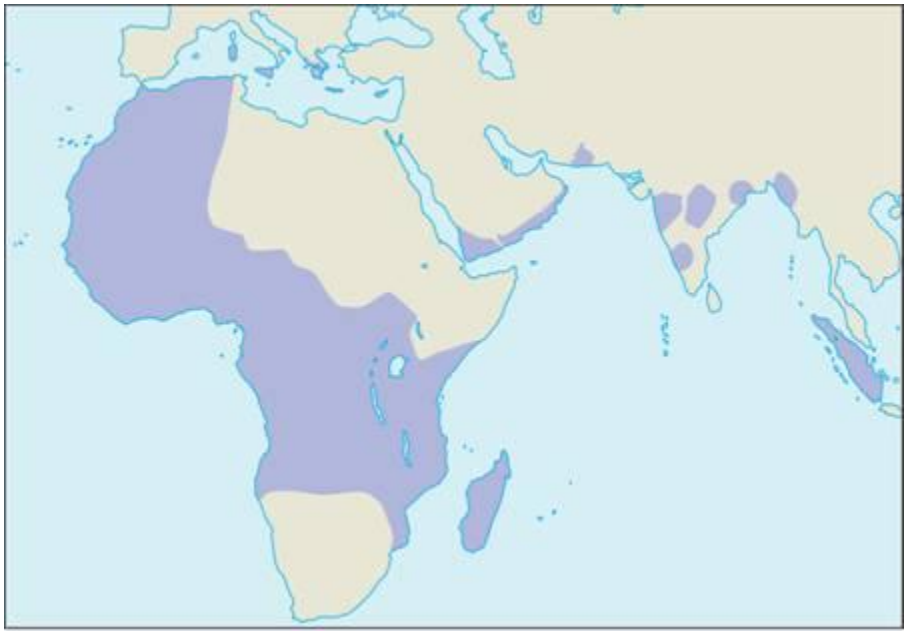
Low oxygen levels cause some red blood cells to become sickle shaped.

People who are heterozygous for the sickle cell allele are generally healthy and they are resistant to malaria.

Malaria and the Sickle Cell Allele



Regions where malaria is common



Regions where the sickle cell allele is common

14–1 Section QUIZ

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Section QUIZ

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ExamView® QuickTake

14–1 Section QUIZ

1 A chromosome that is not a sex chromosome is know as a(an)

A a. autosome.

b. karyotype.

c. pedigree.

d. chromatid.

2 Whether a human will be a male or a female is determined by which

a. sex chromosome is in the egg cell.

b. autosomes are in the egg cell.

A c. sex chromosome is in the sperm cell.

d. autosomes are in the sperm cell.

3 Mendelian inheritance in humans is typically studied by

A a. making inferences from family pedigrees.

b. carrying out carefully controlled crosses.

c. observing the phenotypes of individual humans.

d. observing inheritance patterns in other animals.

4 An individual with a blood type phenotype of O can receive blood from an individual with the phenotype

A a. O.

b. A.

c. AB.

d. B.

5 The ABO blood group is made up of

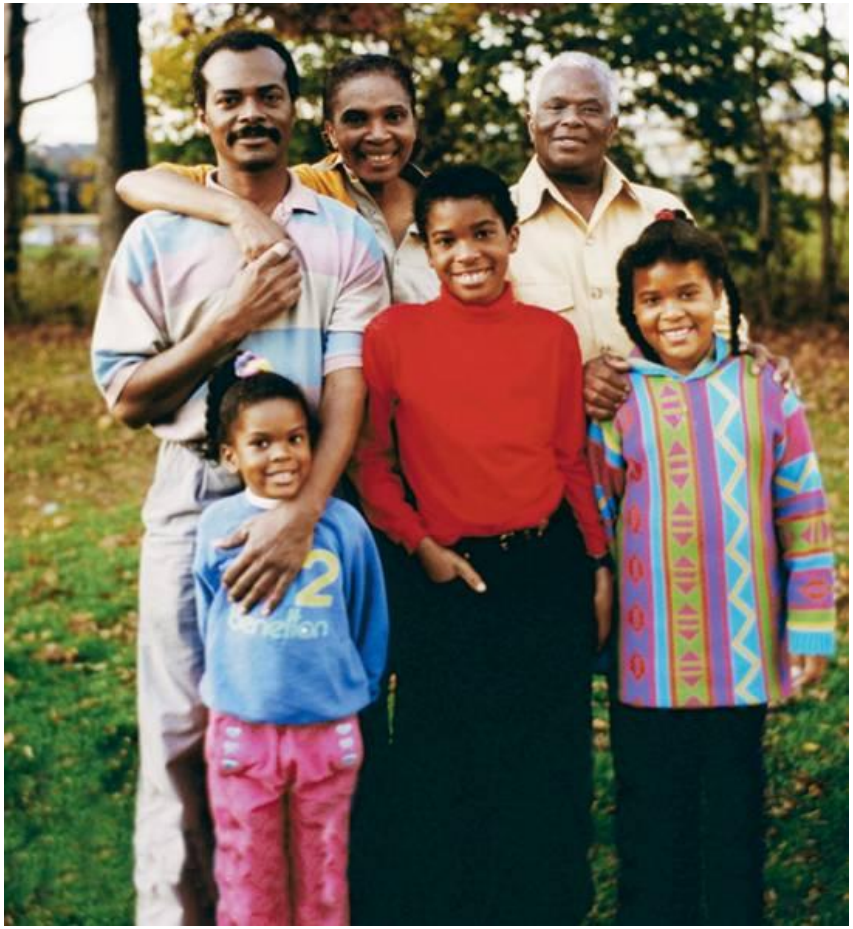
a. two alleles.

A b. three alleles.

c. identical alleles.

d. dominant alleles.

14–2 Human Chromosomes

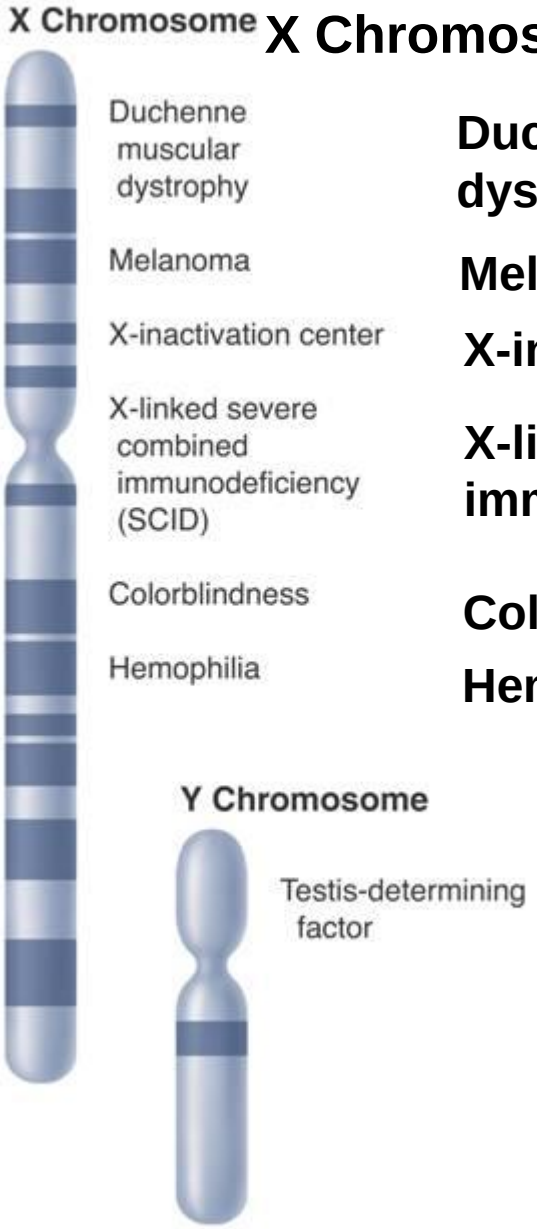


Sex-Linked Genes

a. The X chromosome and the Y chromosomes determine sex.

b. Genes located on these chromosomes are called **sex-linked genes**.

c. More than 100 sex-linked genetic disorders have now been mapped to the X chromosome.



X Chromosome

Duchenne muscular dystrophy

Melanoma

X-inactivation center

X-linked severe combined immunodeficiency (SCID)

Colorblindness

Hemophilia

Y Chromosome

Testis-determining factor

Why are sex-linked disorders more common in males than in females?

For a recessive allele to be expressed in females, there must be two copies of the allele, one on each of the two X chromosomes.

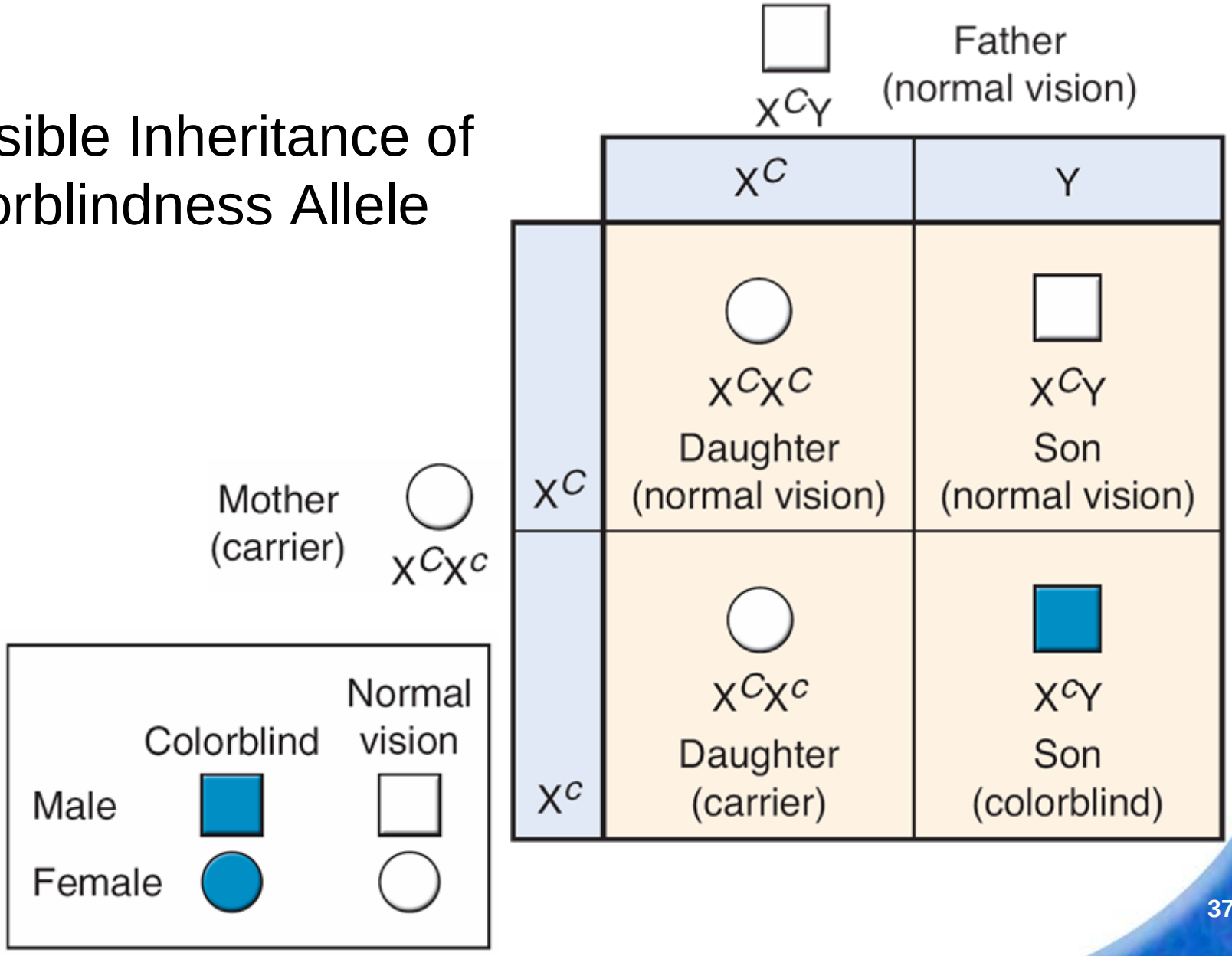
Males have just one X chromosome. Thus, all X-linked alleles are expressed in males, even if they are recessive.

Colorblindness

Three human genes associated with color vision are located on the X chromosome.

In males, a defective version of any one of these genes produces colorblindness.

Possible Inheritance of Colorblindness Allele



Chromosomal Disorders

What problems does nondisjunction cause?

The most common error in meiosis occurs when homologous chromosomes fail to separate.

This is known as **nondisjunction**, which means, “not coming apart.”

If nondisjunction occurs, abnormal numbers of chromosomes may find their way into gametes, and a disorder of chromosome numbers may result.

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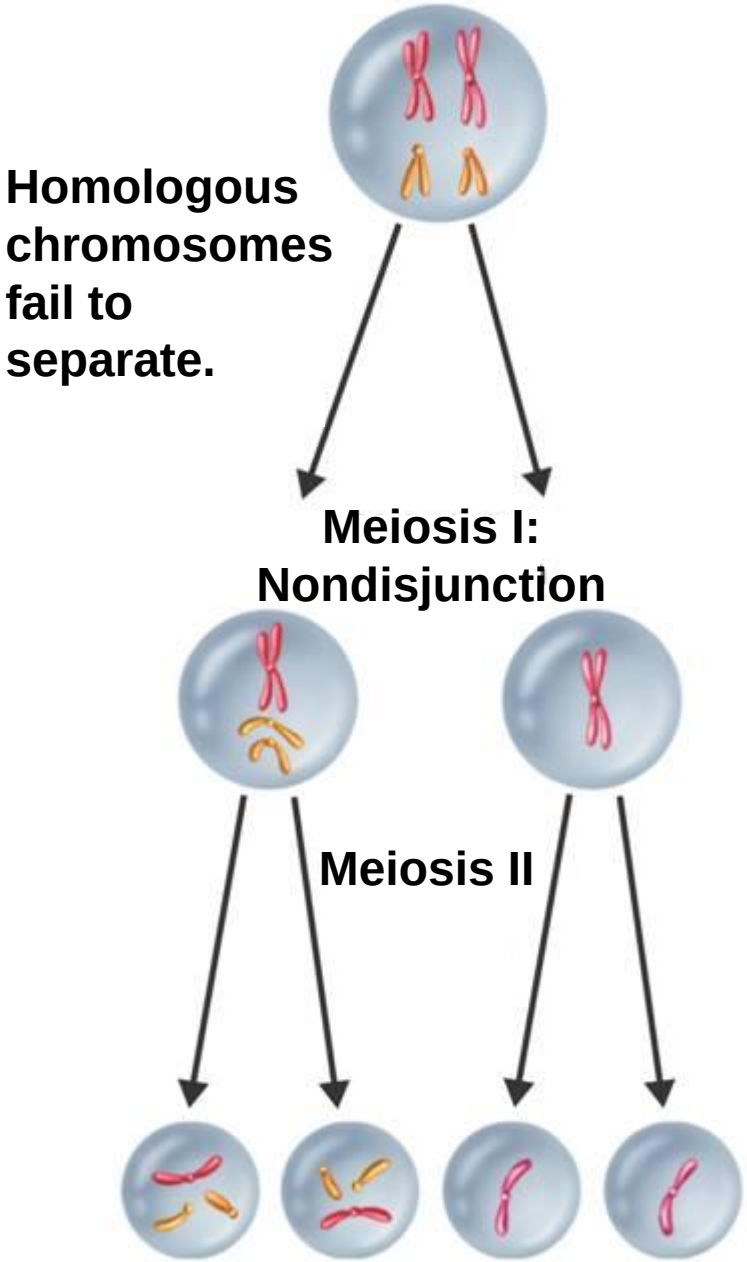
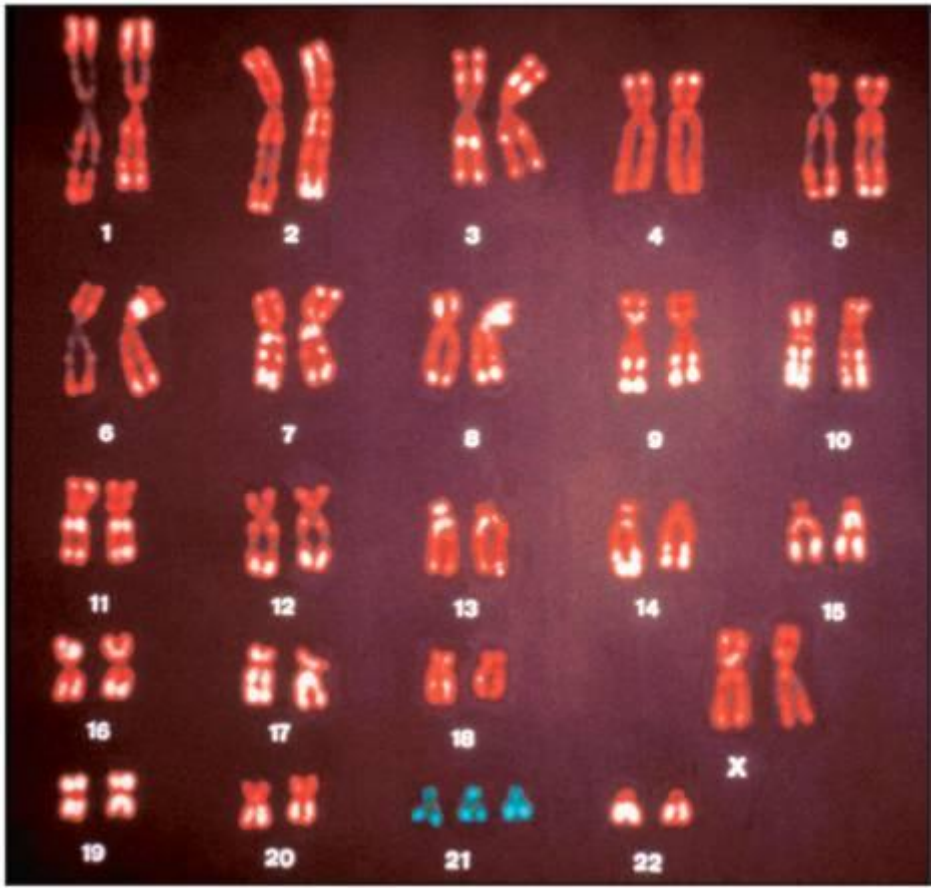
Nondisjunction



Down Syndrome Karyotype



Sex Chromosome Disorders

In females, nondisjunction can lead to Turner's syndrome.

A female with Turner's syndrome usually inherits only one X chromosome (karyotype 45,X).

Women with Turner's syndrome are sterile.

In males, nondisjunction causes Klinefelter's syndrome (karyotype 47,XXY).

The extra X chromosome interferes with meiosis and usually prevents these individuals from reproducing.

14–2 Section QUIZ

Continue to:

Section QUIZ

- or -

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ExamView® QuickTake

14–2 Section QUIZ

1 The average human gene consists of how many base pairs of DNA?

A a. 3000

b. 300

c. 20

d. 30,000

2 Which of the following genotypes indicates an individual who is a carrier for colorblindness?

a. X^CX

A b. X^CX^c

c. X^cY

d. X^CY

3 Colorblindness is much more common in males than in females because

A a. the recessive gene on the male's single X chromosome is expressed.

b. genes on the Y chromosome make genes on the X chromosome more active.

c. females cannot be colorblind.

d. colorblindness is dominant in males and recessive in females.

4 The presence of a dense region in the nucleus of a cell can be used to determine the

A a. sex of an individual.

b. blood type of an individual.

c. chromosome number of an individual.

d. genotype of an individual.

5 Nondisjunction occurs during

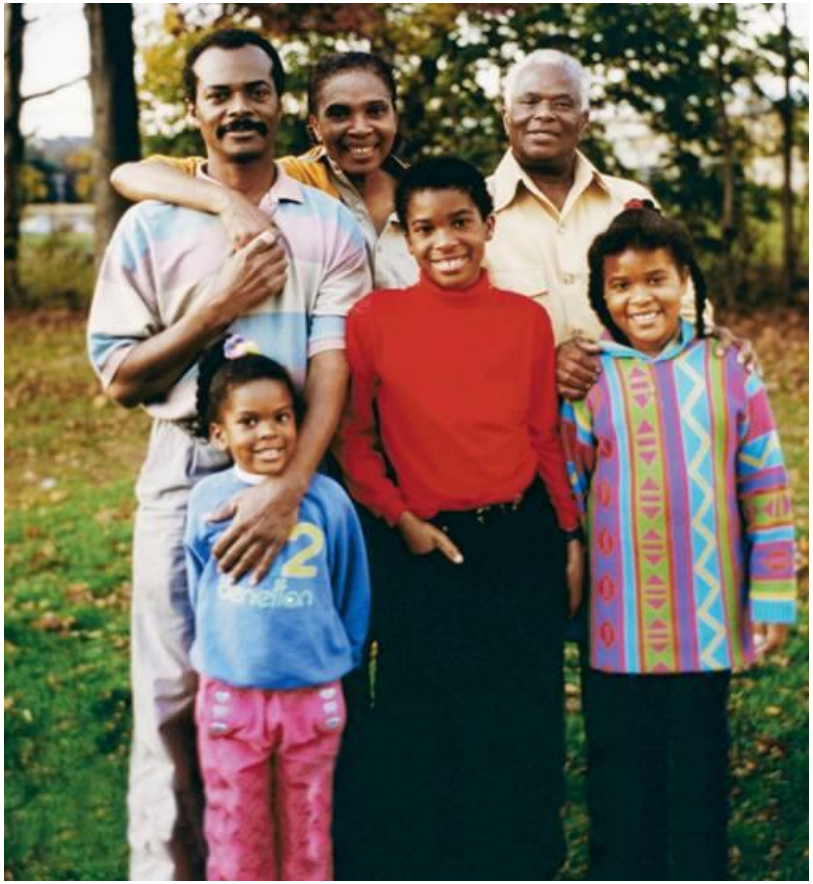
A a. meiosis I.

b. mitosis.

c. meiosis II.

d. between meiosis I and II.

14–3 Human Molecular Genetics



Human DNA Analysis

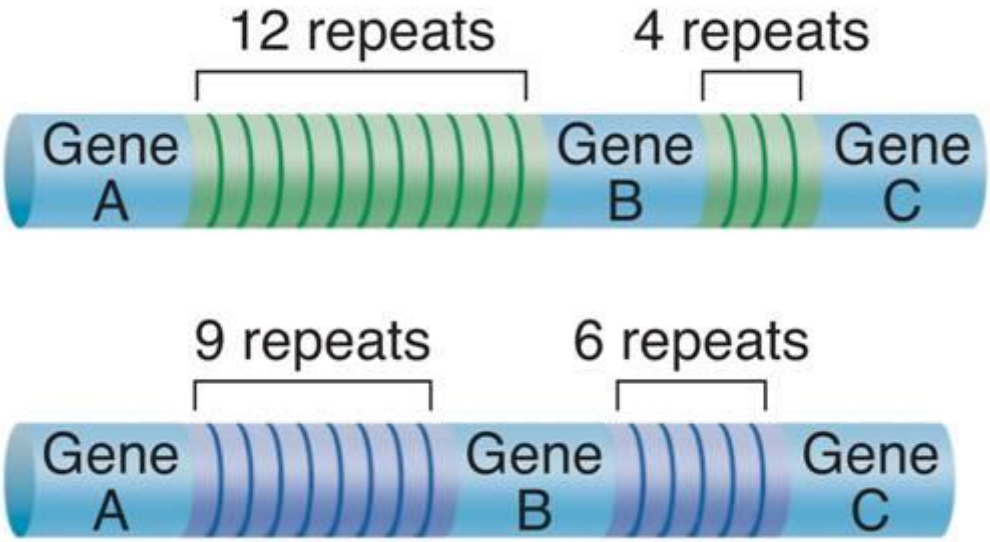
a. There are roughly 6 billion base pairs in your DNA.

b. Biologists search the human genome using sequences of DNA bases.

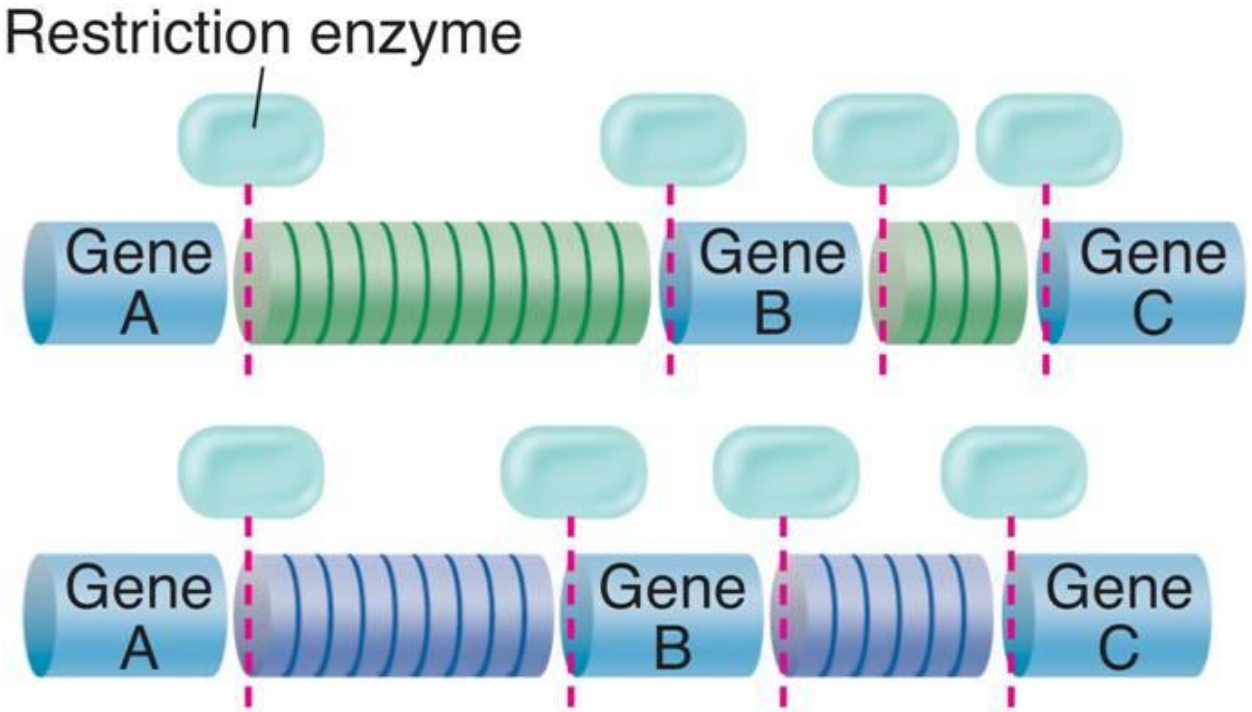
Section QUIZ

Chromosomes contain large amounts of DNA called repeats that do not code for proteins.

This DNA pattern varies from person to person.



Restriction enzymes are used to cut the DNA into fragments containing genes and repeats.

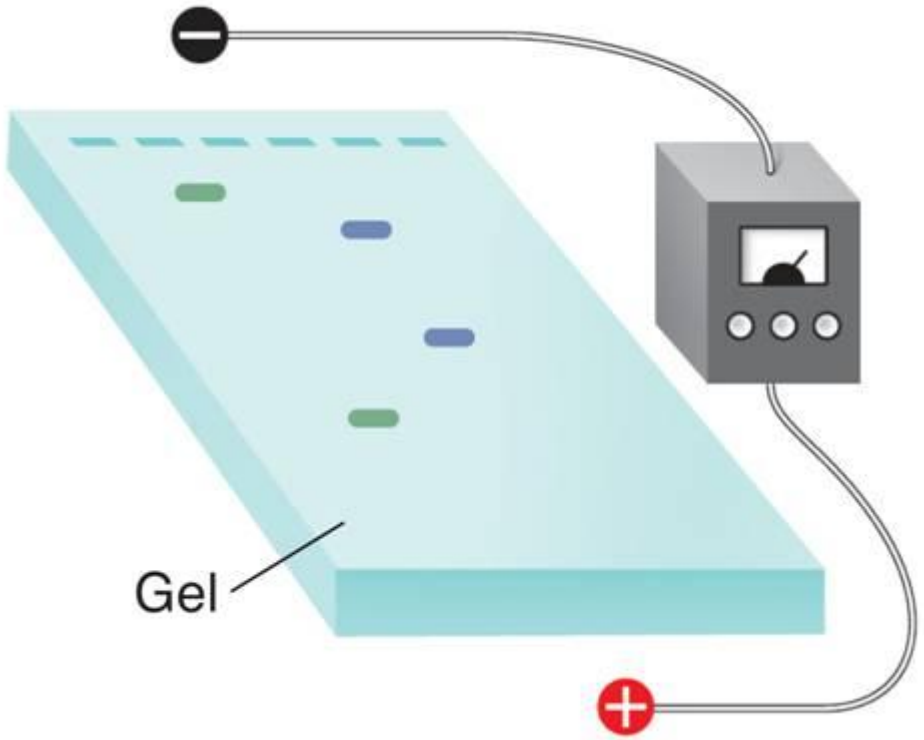


Section QUIZ

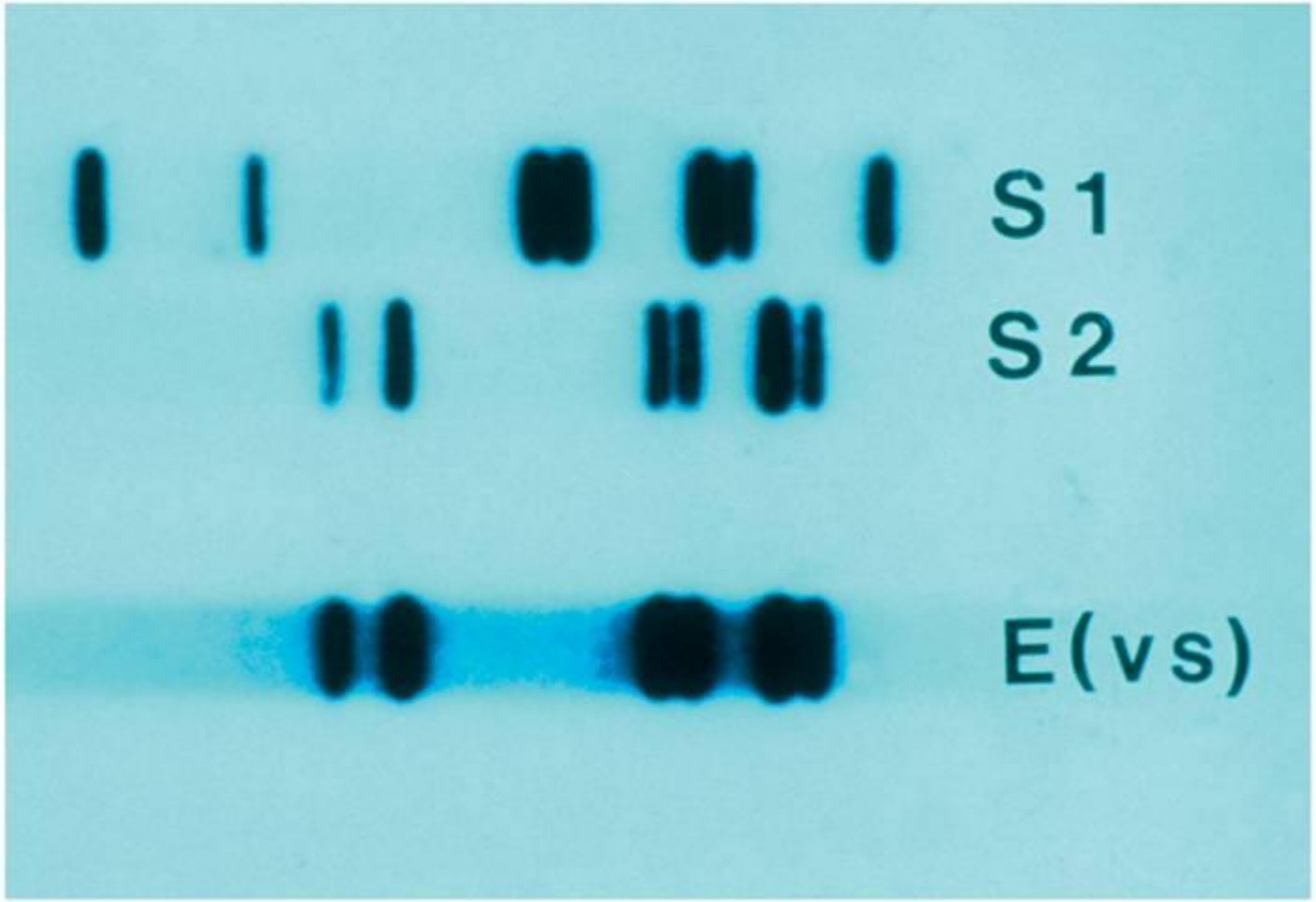
DNA fragments are separated using gel electrophoresis.

Fragments containing repeats are labeled.

This produces a series of bands—the DNA fingerprint.



DNA Fingerprint



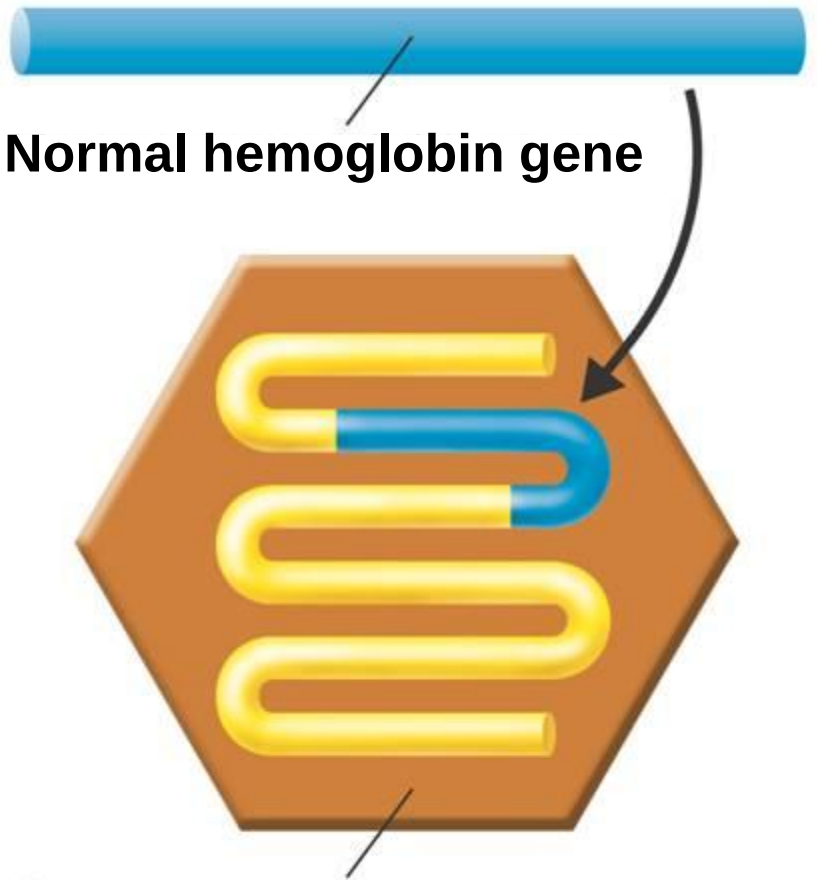
In **gene therapy**, an absent or faulty gene is replaced by a normal, working gene.

The body can then make the correct protein or enzyme, eliminating the cause of the disorder.

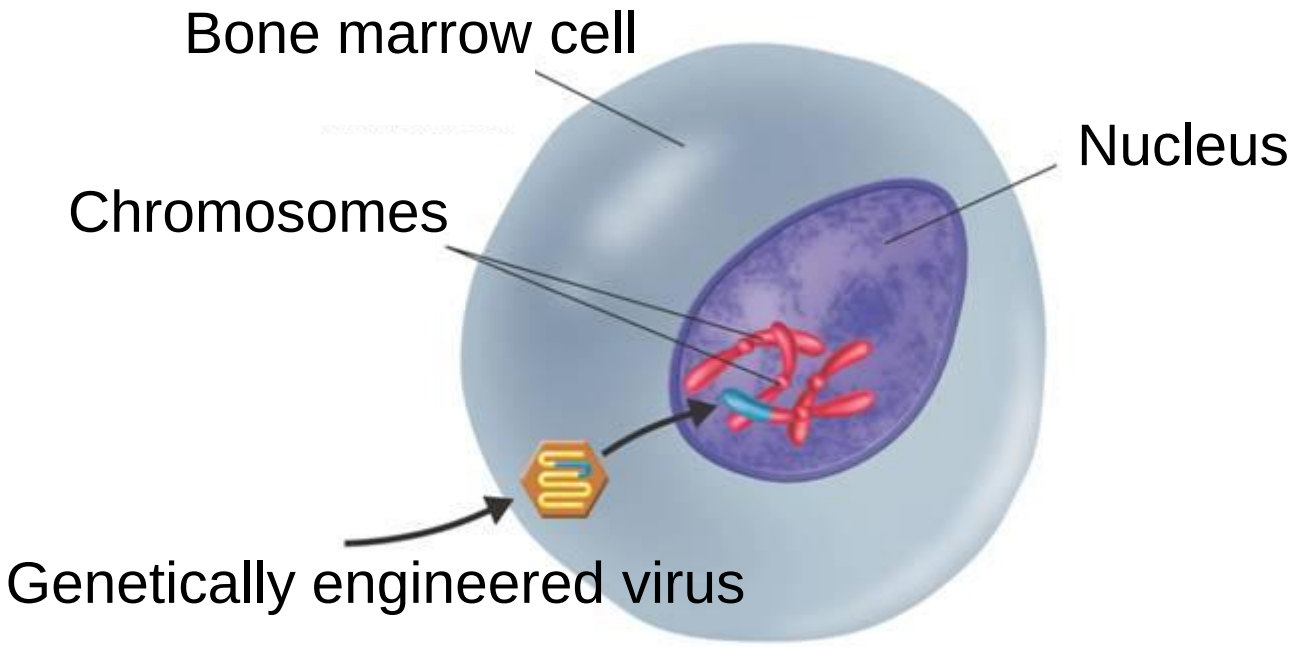
Section QUIZ

Viruses are often used because of their ability to enter a cell's DNA.

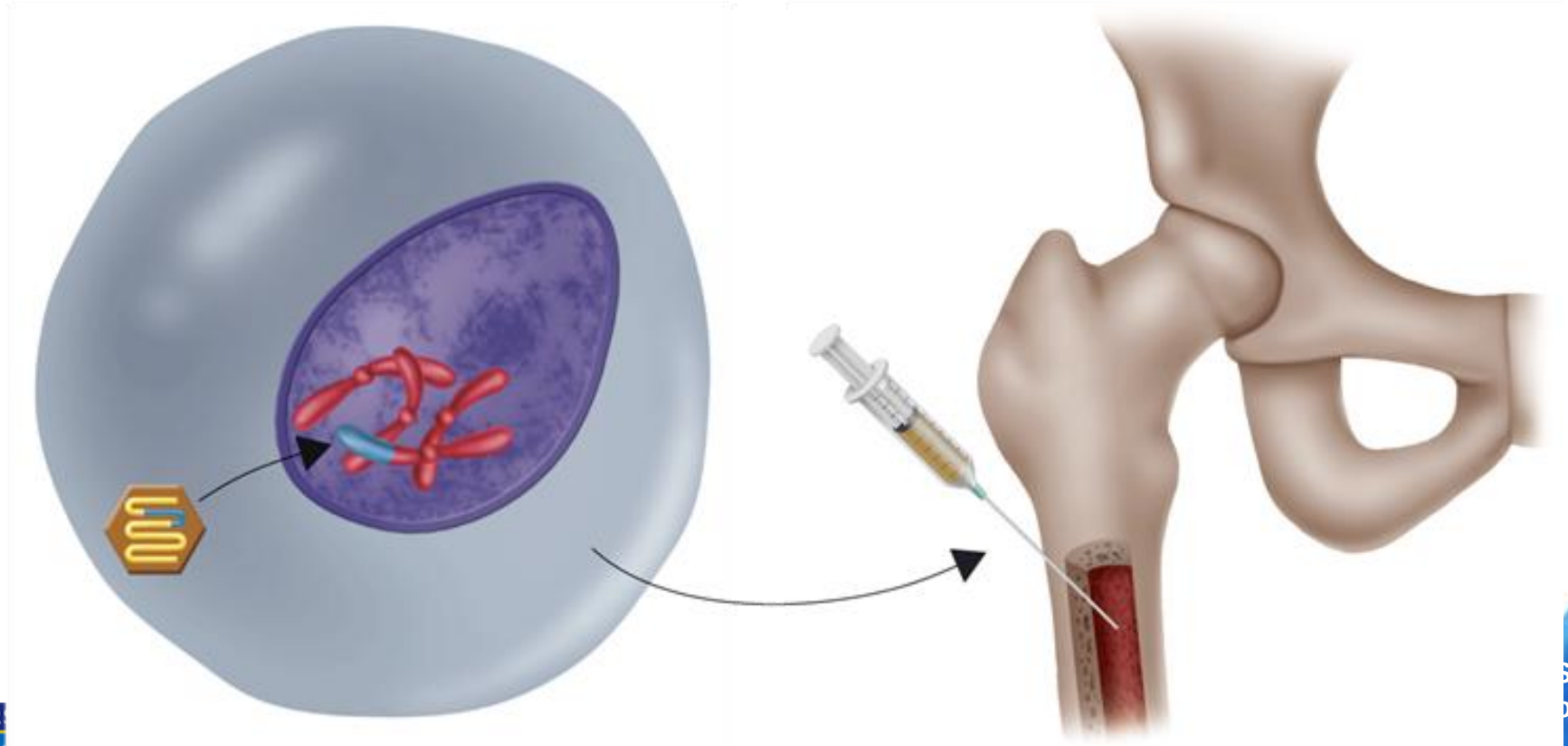
Virus particles are modified so that they cannot cause disease.



A DNA fragment containing a replacement gene is spliced to viral DNA.



The patient is then infected with the modified virus particles, which should carry the gene into cells to correct genetic defects.



14–3 Section QUIZ

1 DNA fingerprinting analyzes sections of DNA that have

a. little or no known function but are identical from one individual to another.

A b. little or no known function but vary widely from one individual to another.

c. a function and are identical from one individual to another.

d. a function and are highly variable from one individual to another.

2 DNA fingerprinting uses the technique of

a. gene therapy.

b. allele analysis.

A c. gel electrophoresis.

d. gene recombination.

3 Repeats are areas of DNA that

A a. do not code for proteins.

b. code for proteins.

c. are identical from person to person.

d. cause genetic disorders.

4 Data from the human genome project is available

a. only to those who have sequenced the DNA.

b. to scientists who are able to understand the data.

c. by permission to anyone who wishes to do research.

A d. to anyone with Internet access.

5 Which statement most accurately describes gene therapy?

a. It repairs the defective gene in all cells of the body.

b. It destroys the defective gene in cells where it exists.

A c. It replaces absent or defective genes with a normal gene.

d. It promotes DNA repair through the use of enzymes.

END OF SECTION