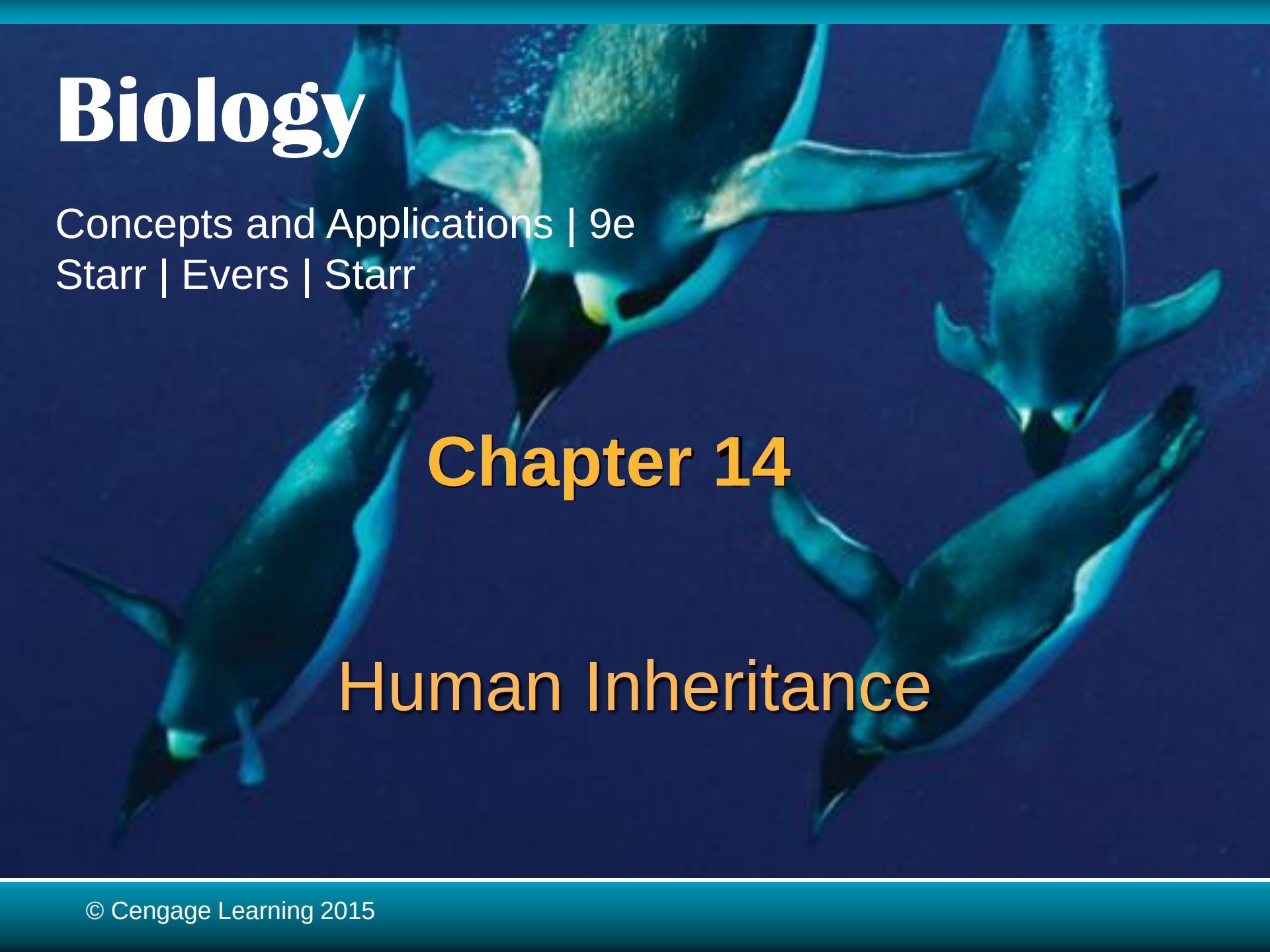

Biology

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Chapter 14

Human Inheritance

14.1 How Do We Study Inheritance Patterns In Humans?

- Pea plants and fruit flies are perfect for genetic studies because:
  - They have few chromosomes
  - They reproduce quickly
  - They can survive in lab conditions
  - There are few ethical problems with their use

How Do We Study Inheritance Patterns In Humans? (cont'd.)

- Humans involve more issues
  - Geneticists often use historical records
  - Make charts (pedigrees of genetic connections)
    - Allows geneticists to determine the probability that a trait will recur in future generations

How Do We Study Inheritance Patterns In Humans? (cont'd.)

- Types of genetic mutations
  - Single genes that follow Mendelian inheritance patterns govern more than 6,000 genetic abnormalities and disorders
  - Most human traits are *polygenic*, or influenced by multiple genes
  - Traits can be influenced by environmental factors
  - Alleles that give rise to severe genetic disorders are rare

How Do We Study Inheritance Patterns In Humans? (cont'd.)

- Six main patterns of inheritance for genetic abnormalities and disorders:
  - Autosomal dominant inheritance pattern
  - Autosomal recessive inheritance pattern
  - X-linked recessive inheritance pattern
  - X-linked dominant inheritance pattern
  - Changes in chromosome number
  - Changes in chromosome structure

How Do We Study Inheritance Patterns In Humans? (cont'd.)

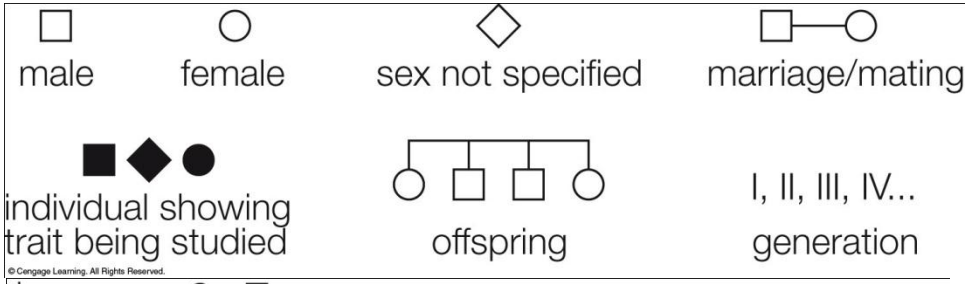
TABLE 14.1

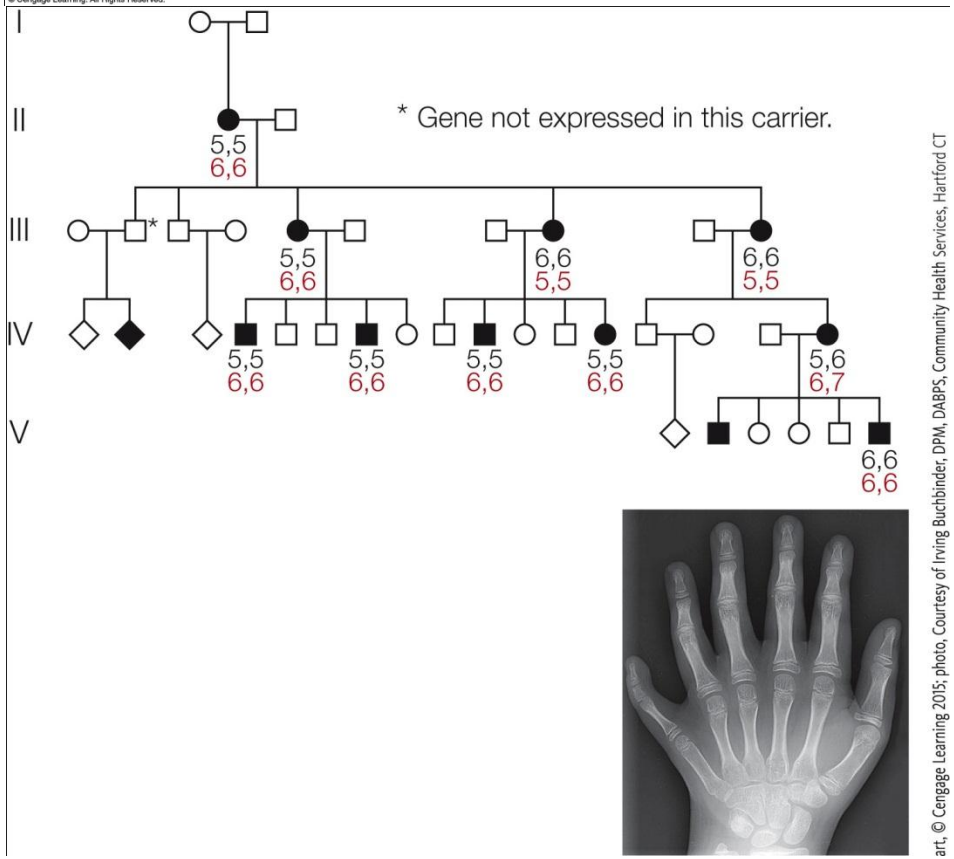
Examples of Genetic Abnormalities and Disorders in Humans

Disorder or Abnormality	Main Symptoms
Autosomal dominant inheritance pattern	
Achondroplasia	One form of dwarfism
Aniridia	Defects of the eyes
Camptodactyly	Rigid, bent fingers
Familial hypercholesterolemia	High cholesterol level; clogged arteries
Huntington's disease	Degeneration of the nervous system
Marfan syndrome	Abnormal or missing connective tissue
Polydactyly	Extra fingers, toes, or both
Progeria	Drastic premature aging
Neurofibromatosis	Tumors of nervous system, skin
Autosomal recessive inheritance pattern	
Albinism	Absence of pigmentation
Hereditary methemoglobinemia	Blue skin coloration
Cystic fibrosis	Difficulty breathing; chronic lung infections
Ellis–van Creveld syndrome	Dwarfism, heart defects, polydactyly
Fanconi anemia	Physical abnormalities, marrow failure
Galactosemia	Brain, liver, eye damage
Hereditary hemochromatosis	Joints, organs damaged by iron overload
Phenylketonuria (PKU)	Mental impairment
Sickle-cell anemia	Anemia, pain, swelling, frequent infections
Tay–Sachs disease	Deterioration of mental and physical abilities; early death
X-linked recessive inheritance pattern	
Androgen insensitivity syndrome	XY individual but having some female traits; sterility
Red–green color blindness	Inability to distinguish red from green
Hemophilia	Impaired blood clotting ability
Muscular dystrophies	Progressive loss of muscle function
X-linked anhidrotic dysplasia	Mosaic skin (patches with or without sweat glands); other ill effects
X-linked dominant inheritance pattern	
Fragile X syndrome	Intellectual, emotional disability
Incontinentia pigmenti	Abnormalities of skin, hair, teeth, nails, eyes; neurological problems
Changes in chromosome number	
Down syndrome	Mental impairment; heart defects
Turner syndrome (XO)	Sterility; abnormal ovaries, sexual traits
Klinefelter syndrome	Sterility; mild mental impairment
XXX syndrome	Minimal abnormalities
XYY condition	Mild mental impairment or no effect
Changes in chromosome structure	
Chronic myelogenous leukemia (CML)	Overproduction of white blood cells; organ malfunctions
Cri-du-chat syndrome	Mental impairment; abnormal larynx

How Do We Study Inheritance Patterns In Humans? (cont'd.)

A





B

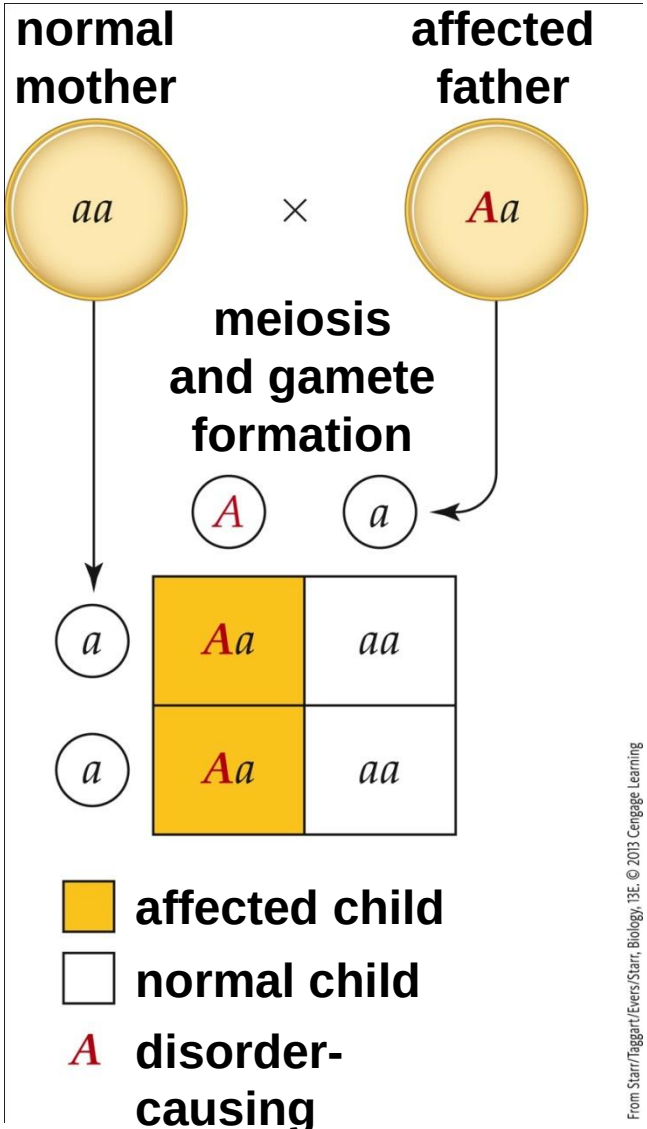
14.2 How Do We Know When A Trait Is Affected By An Allele On An Autosome?

- An allele is inherited in an:
  - Autosomal dominant pattern if the trait it specifies appears in homozygous and heterozygous people
  - Autosomal recessive pattern if the trait it specifies appears only in homozygous people

How Do We Know When A Trait Is Affected By An Allele On An Autosome? (cont'd.)

- The autosomal dominant pattern
  - An autosomal dominant trait appears in every generation
  - When one parent is heterozygous, and the other is homozygous recessive, each child has a 50% chance of inheriting the dominant allele and displaying the trait

How Do We Know When A Trait Is Affected By An Allele On An Autosome? (cont'd.)



How Do We Know When A Trait Is Affected By An Allele On An Autosome? (cont'd.)

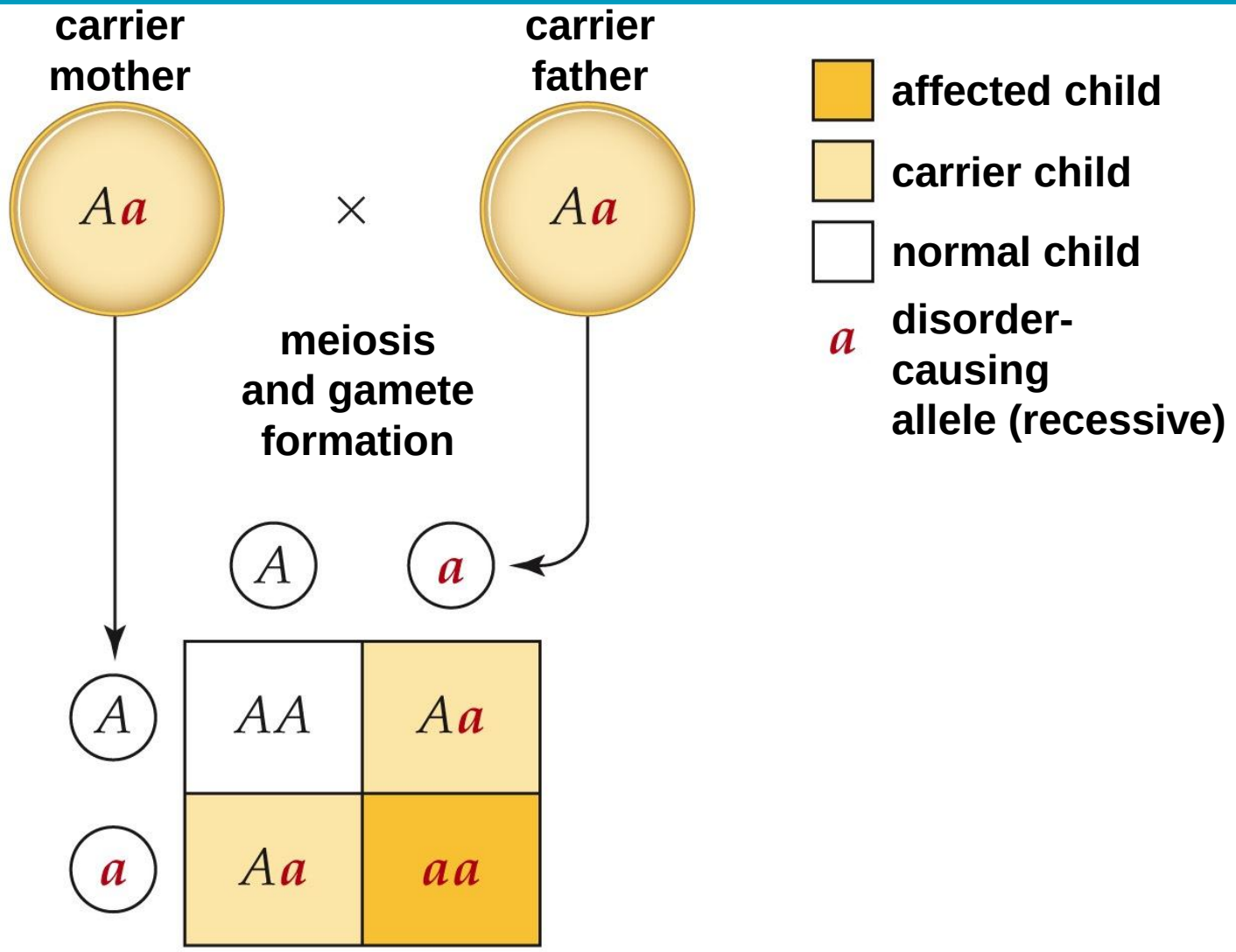
- Autosomal dominant disorders

Disorder	Main Symptoms
Achondroplasia	One form of dwarfism
Aniridia	Defects of the eyes
Camptodactyly	Rigid, bent fingers
Hypercholesterolemia	High cholesterol level
Huntington's disease	Degeneration of nervous system
Marfan syndrome	Abnormal connective tissue
Polydactyly	Extra fingers, toes, or both
Progeria	Drastic premature aging
Neurofibromatosis	Tumors of nervous system, skin

How Do We Know When A Trait Is Affected By An Allele On An Autosome? (cont'd.)

- Autosomal recessive pattern
  - An autosomal allele is inherited in a recessive pattern if it is expressed only in homozygous people, so recessive traits may skip generations
  - People heterozygous for the allele are carriers; they have the allele but not the trait
  - Each child of two carriers has a 25% chance of being homozygous and having the trait

How Do We Know When A Trait Is Affected By An Allele On An Autosome? (cont'd.)



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How Do We Know When A Trait Is Affected By An Allele On An Autosome? (cont'd.)

Disorder	Main Symptoms
Albinism	Absence of pigmentation
Methemoglobinemia	Blue skin coloration
Cystic fibrosis	Abnormal glandular secretions leading to tissue and organ damage
Ellis–van Creveld	Dwarfism, heart defects, polydactyly
Fanconi anemia	Abnormalities, bone marrow failure
Galactosemia	Brain, liver, eye damage
Hemochromatosis	Iron overload , joint & organ damage
Phenylketonuria (PKU)	Mental impairment
Sickle-cell anemia	Adverse pleiotropic effects
Tay–Sachs disease	Deterioration of mental and physical abilities; early death

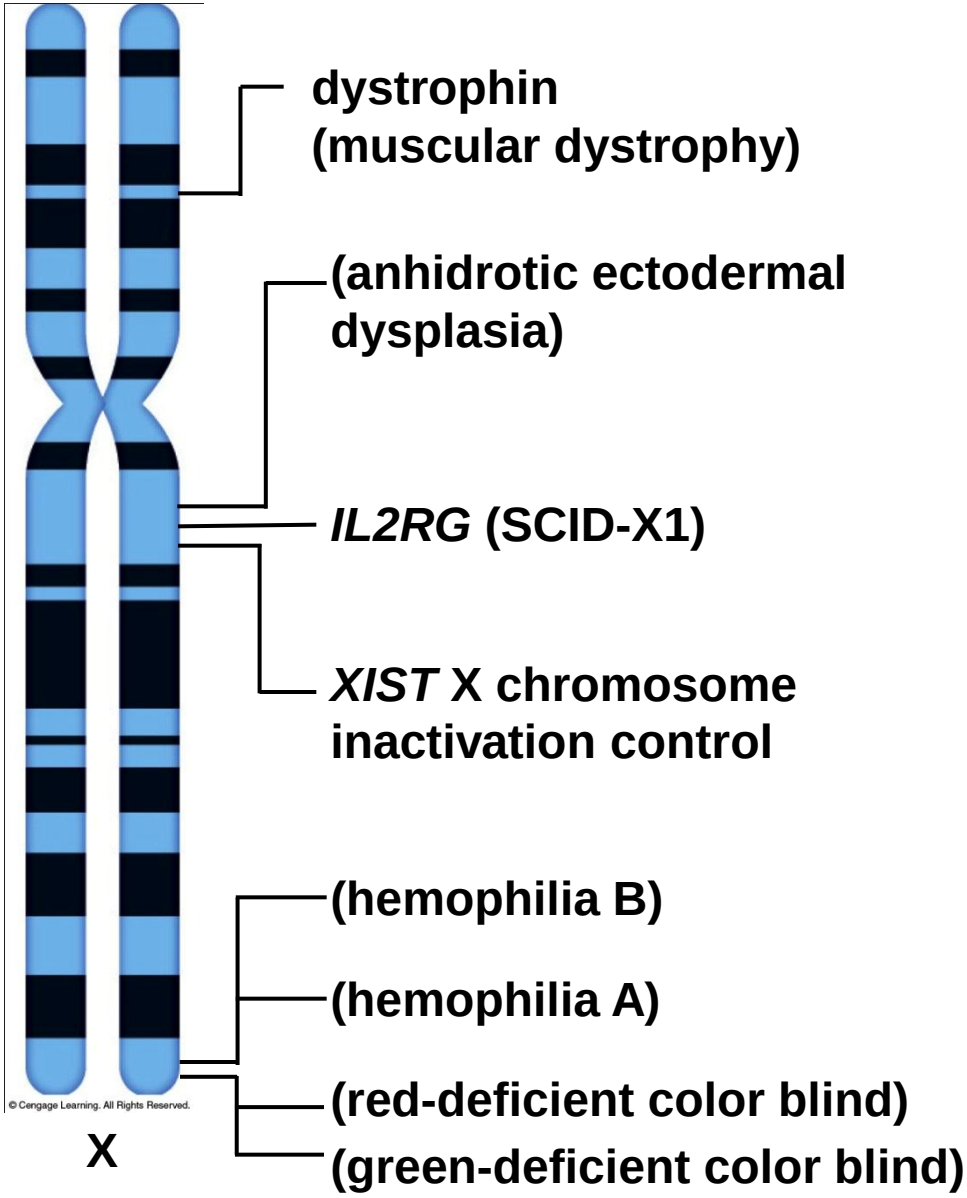
14.3 How Do We Know When A Trait Is Affected By An Allele On An X Chromosome?

- X-linked recessive pattern
  - An allele is inherited on the X chromosome
  - Most are recessive, because X-linked dominant alleles tend to be lethal in male embryos

How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

- X-linked recessive pattern (cont'd.)
  - X-linked recessive disorders tend to appear in men more often than in women
    - Men (XY) have only one X chromosome
    - Women have two X chromosomes (XX), so they can be heterozygous for a recessive allele
  - Men can transmit an X-linked allele to daughters, but not to sons – only a woman can pass an X-linked allele to a son

How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

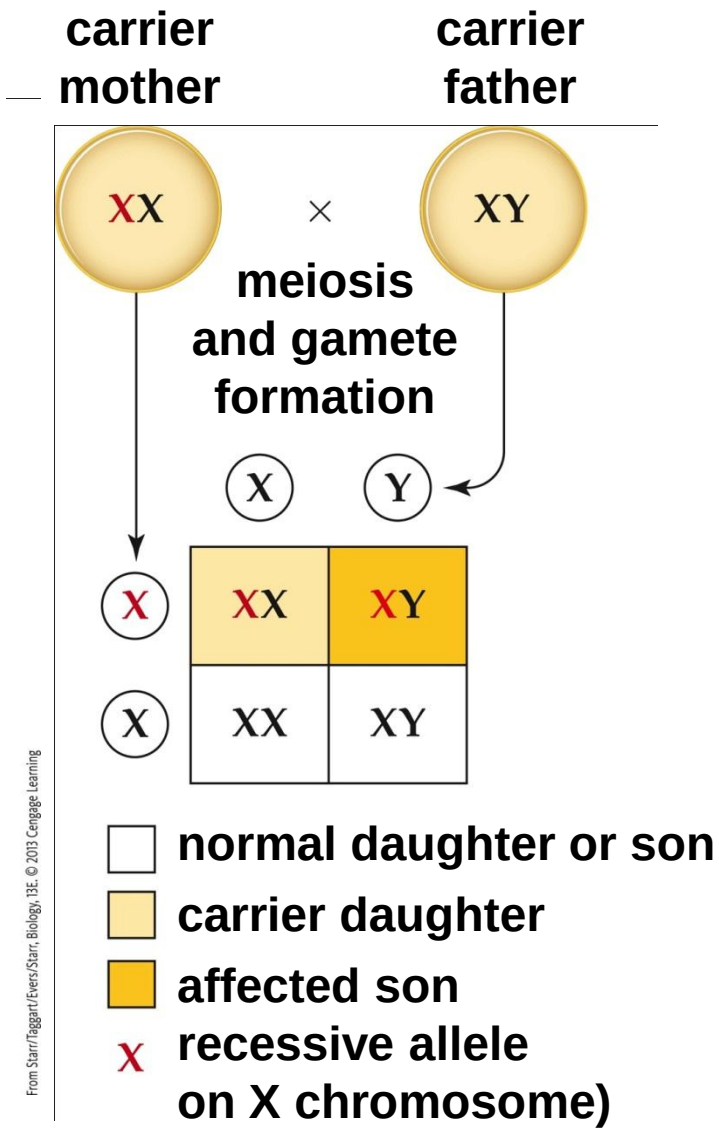


How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

- X- linked recessive disorders

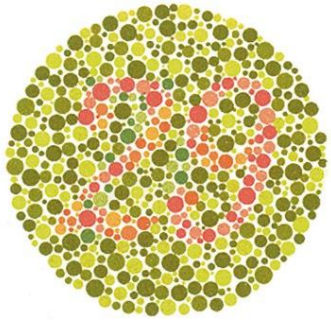
Disorder	Main Symptoms
Androgen insensitivity syndrome	XY individual but having some female traits; sterility
Red-green color blindness	Inability to distinguish red from green
Hemophilia	Impaired blood clotting ability
Muscular dystrophies	Progressive loss of muscle function
X-linked anhidrotic dysplasia	Mosaic skin (patches with or without sweat glands); other effects

How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)



How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

left, Gary L. Friedman, www.FriedmanArchives.com.; right, Life Nature Library, The Primates, 1965, Sarel Eimerl and Irven DeVore.



You may have one form of red–green color blindness if you see a 7 in this circle instead of a 29.

You may have another form of red–green color blindness if you see a 3 instead of an 8 in this circle.

How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

- Most genes involved in proper function of pigment-containing receptors in the eyes are on the X chromosome
  - Color blindness includes a range of conditions in which an individual cannot distinguish among some or all colors
    - Some confuse red and green colors; others see green as shades of gray, but perceive blues and yellows quite well

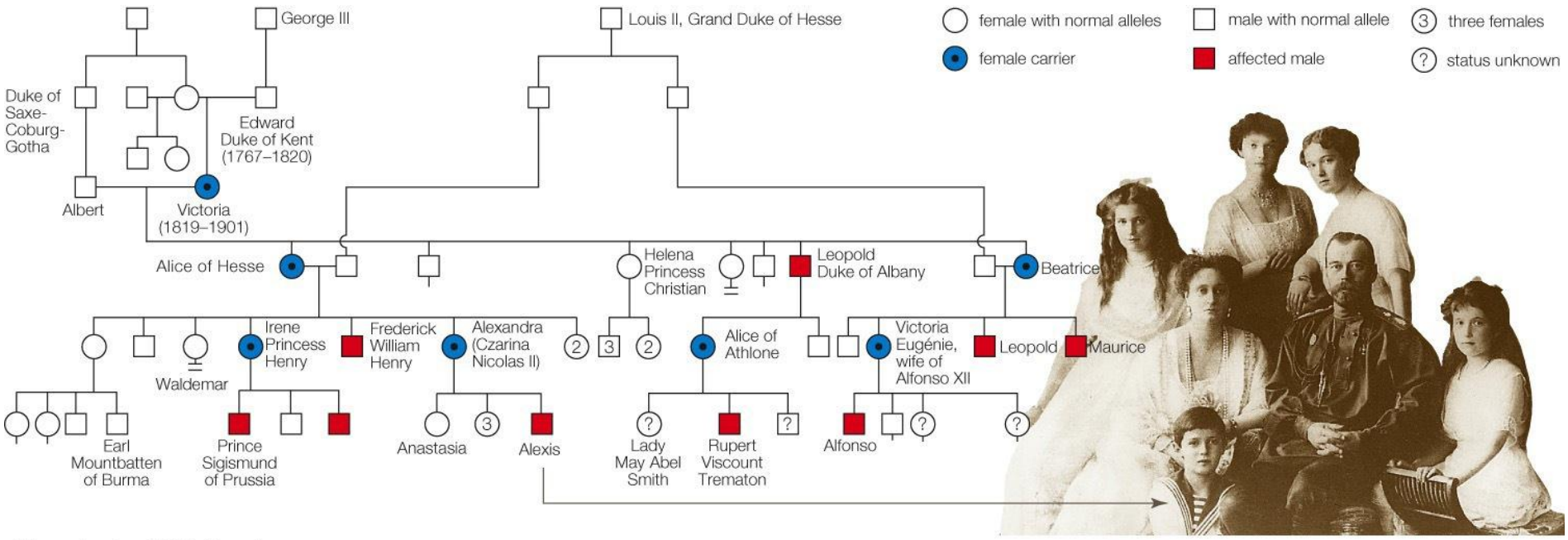
How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

- Duchenne muscular dystrophy (DMD)
  - X-linked recessive disorder
  - Causes the protein, dystrophin to be absent
  - Muscle and nerve cells become replaced by fat cells
  - Affects 1/3,500 people, mostly boys
  - Boys with DMD are in a wheelchair by age 12, and die from a heart disorder or respiratory failure before age 30

How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)

- Hemophilia A
  - X-linked recessive disorder
  - Interferes with blood clotting
  - Involves factor VIII
  - In the 19th century, it was relatively common in royal families of Europe and Russia

How Do We Know When A Trait Is Affected By An Allele On An X Chromosome? (cont'd.)



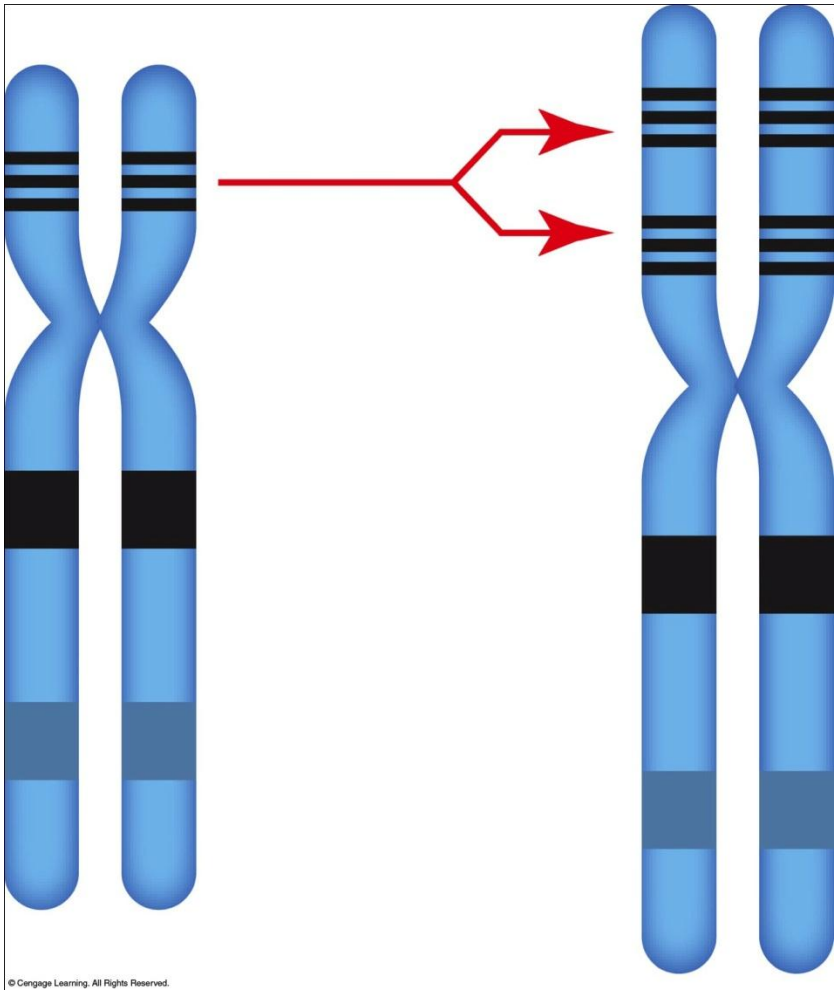
14.4 How Does Chromosome Structure Change?

- *Mutations* are small-scale changes in DNA sequence
- Chromosome mutations
  - Include duplications, deletions, inversions, and translocations
  - Have been evolutionarily important
  - Tend to result in genetic disorders

How Does Chromosome Structure Change? (cont'd.)

- Duplications
  - Repeated section of a chromosome
  - Occur during prophase I of meiosis
  - Cause genetic abnormalities or disorders
    - Huntington's disease

How Does Chromosome Structure Change? (cont'd.)

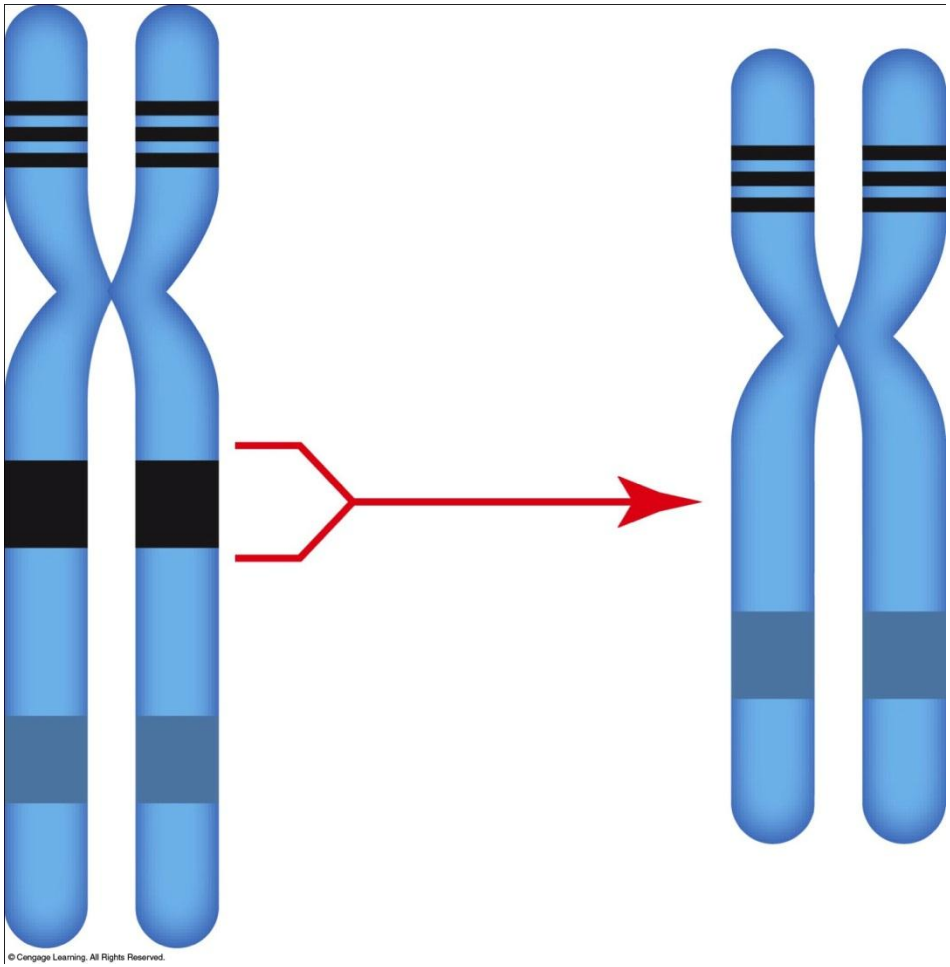


A Duplication

How Does Chromosome Structure Change? (cont'd.)

- Deletions
  - Loss of a part of a chromosome
  - In mammals can cause serious disorders and are often lethal
  - Examples include Duchenne muscular dystrophy and cri-du-chat

How Does Chromosome Structure Change? (cont'd.)

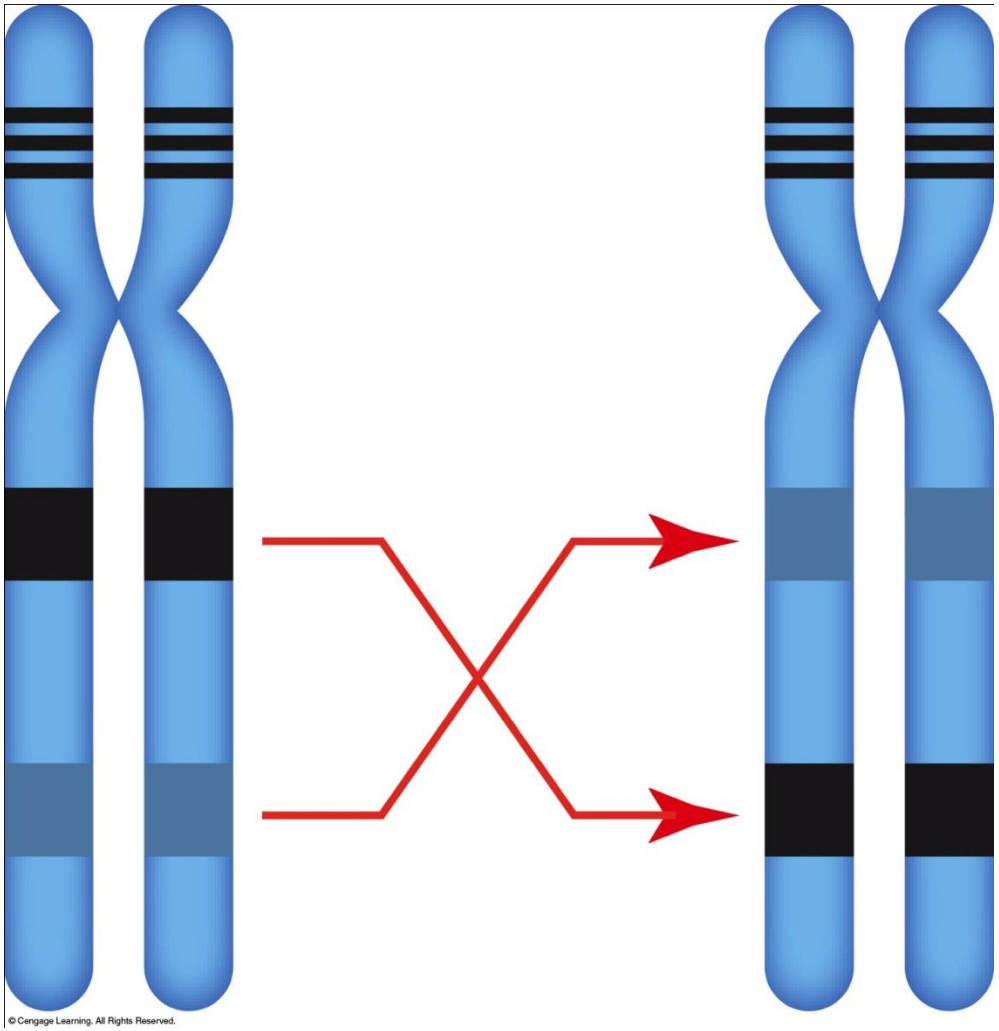


B Deletion

How Does Chromosome Structure Change? (cont'd.)

- Inversion
  - Structural rearrangement of a chromosome
  - Part becomes oriented in the reverse direction
  - No molecular loss
  - May not affect carrier's health
  - May effect fertility
  - Can produce other abnormalities that reduce viability of embryos

How Does Chromosome Structure Change? (cont'd.)

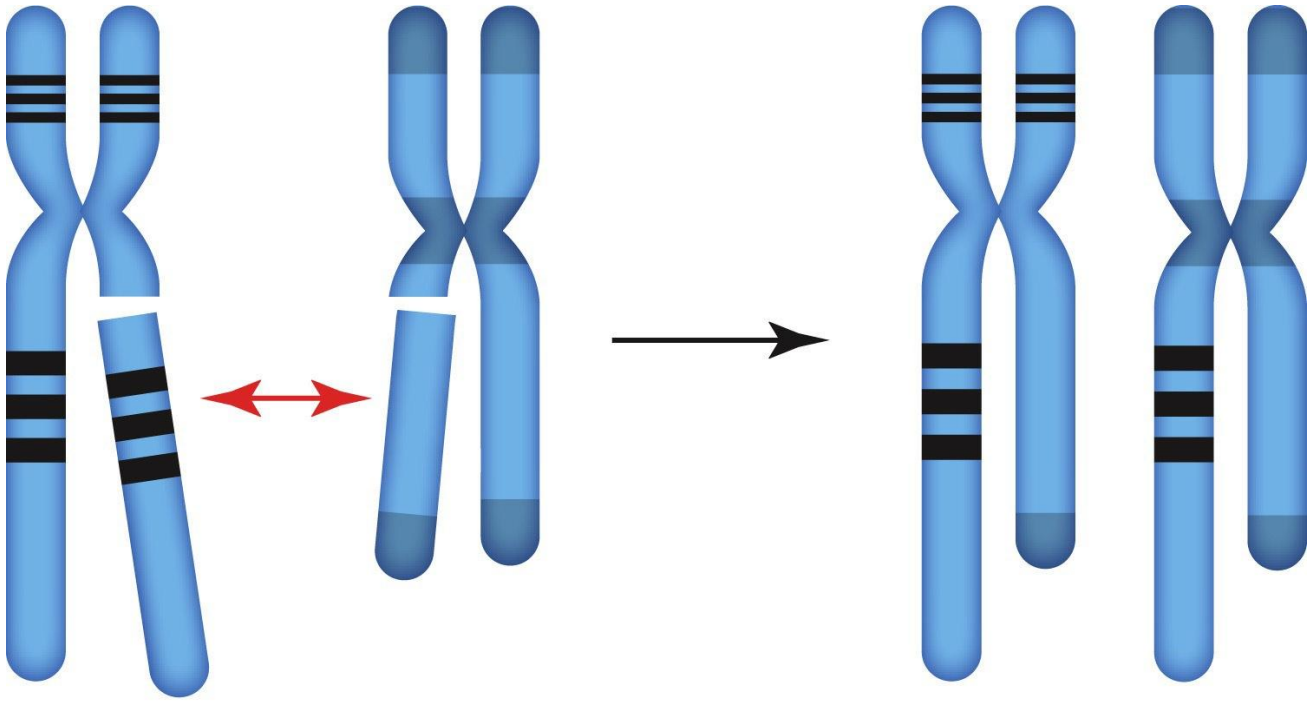


C Inversion

How Does Chromosome Structure Change? (cont'd.)

- Translocation of a chromosome
  - A broken piece gets reattached in the wrong location
  - The broken part may attach to a different chromosome, or to a different part of the same one
  - Most translocations are reciprocal, or balanced; two chromosomes exchange broken parts
  - Can affect fertility

How Does Chromosome Structure Change? (cont'd.)



D Translocation

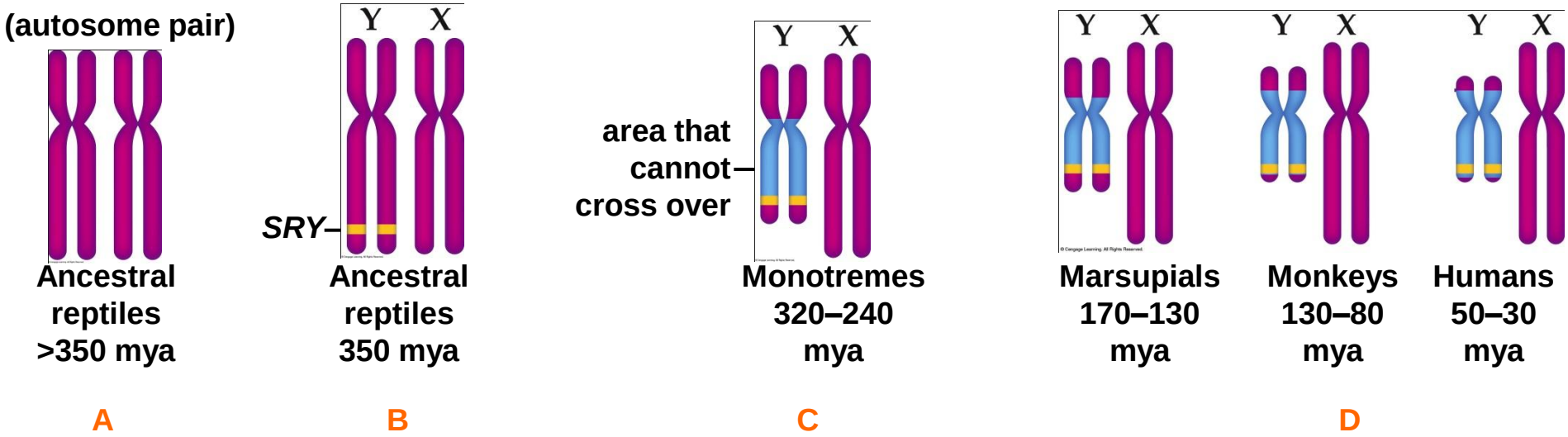
How Does Chromosome Structure Change? (cont'd.)

- Chromosome changes in evolution
  - Most major alterations are harmful or lethal in humans
  - Many major structural changes have accumulated in chromosomes of all species over evolutionary time
  - Speciation occurs by large-scale changes in chromosomes

How Does Chromosome Structure Change? (cont'd.)

- Evolution of the Y chromosome
  - X and Y chromosomes were once homologous autosomes
  - About 350 mya, a gene on one chromosome mutated – interfering with crossing over during meiosis – and mutations began to accumulate separately in the two chromosomes
  - Today, the SRY gene (Y chromosome) determines male sex

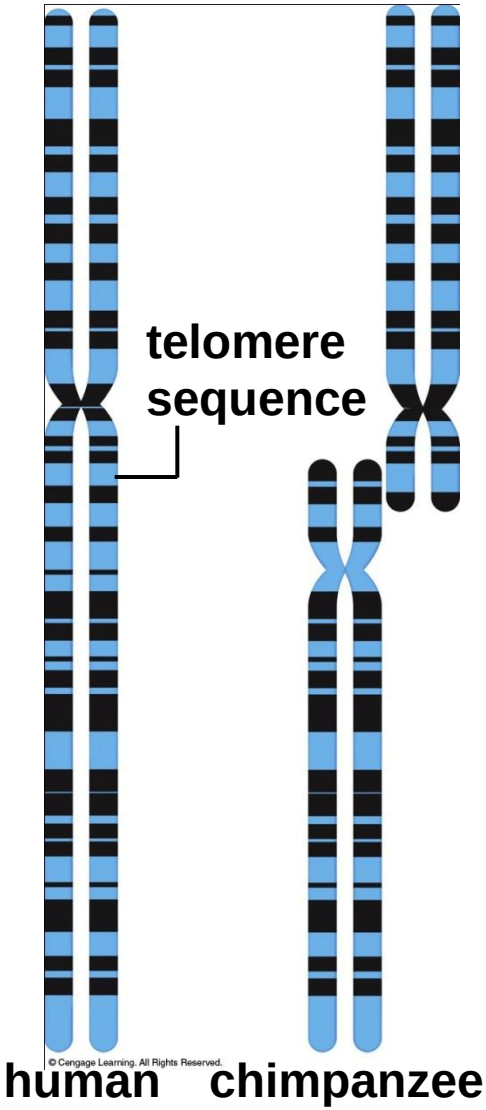
How Does Chromosome Structure Change? (cont'd.)



How Does Chromosome Structure Change? (cont'd.)

- Human somatic cells have 23 pairs of chromosomes
- Chimpanzees, gorillas, and orangutans have 24 pairs
- During human evolution, two chromosomes fused end to end and formed our chromosome 2

How Does Chromosome Structure Change? (cont'd.)



14.5 What Are The Effects of Chromosome Number Changes in Humans?

- Occasionally abnormal events can occur
  - Happens before or during meiosis
  - New individuals end up with the wrong chromosome number
  - Consequences range from minor to lethal changes in form and function

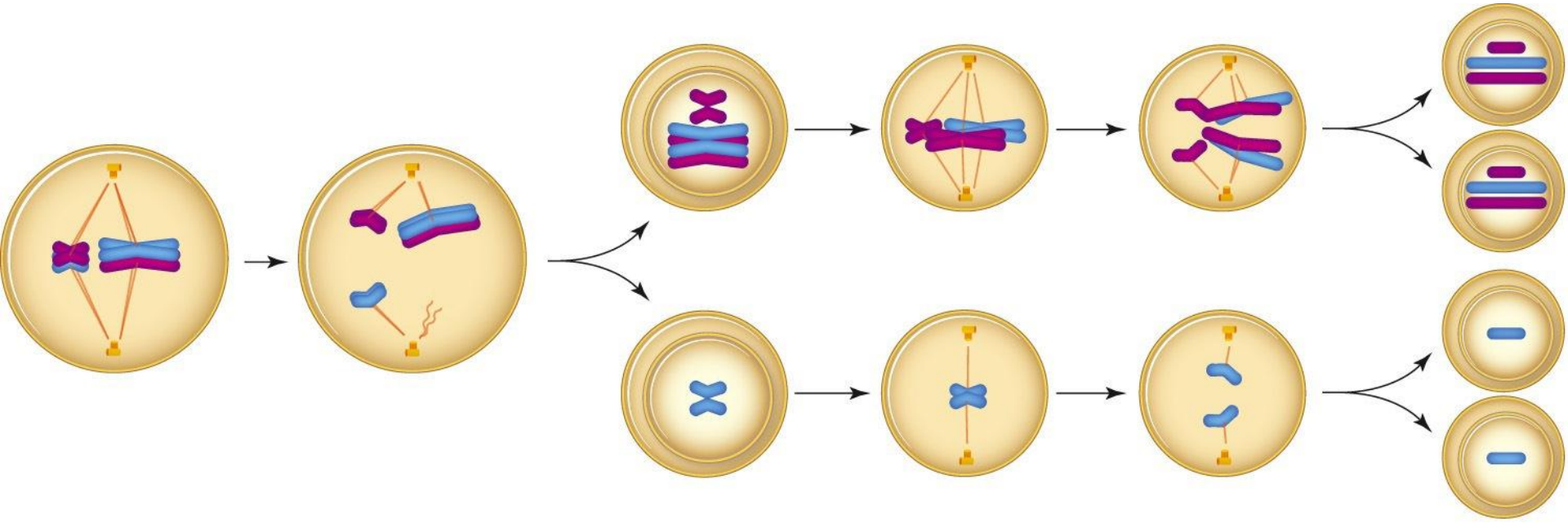
What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Polyploidy
  - Individuals have three or more of each type of chromosome
  - Lethal in humans
  - Many flowering plants, and some insects, fishes, and other animals, are polyploid

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Nondisjunction
  - Failure of sister chromatids or homologous chromosomes to separate during nuclear division
  - Changes in chromosome number are usually caused by nondisjunction
  - Affects chromosome number at fertilization
  - Causes genetic disorders among resulting offspring

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)



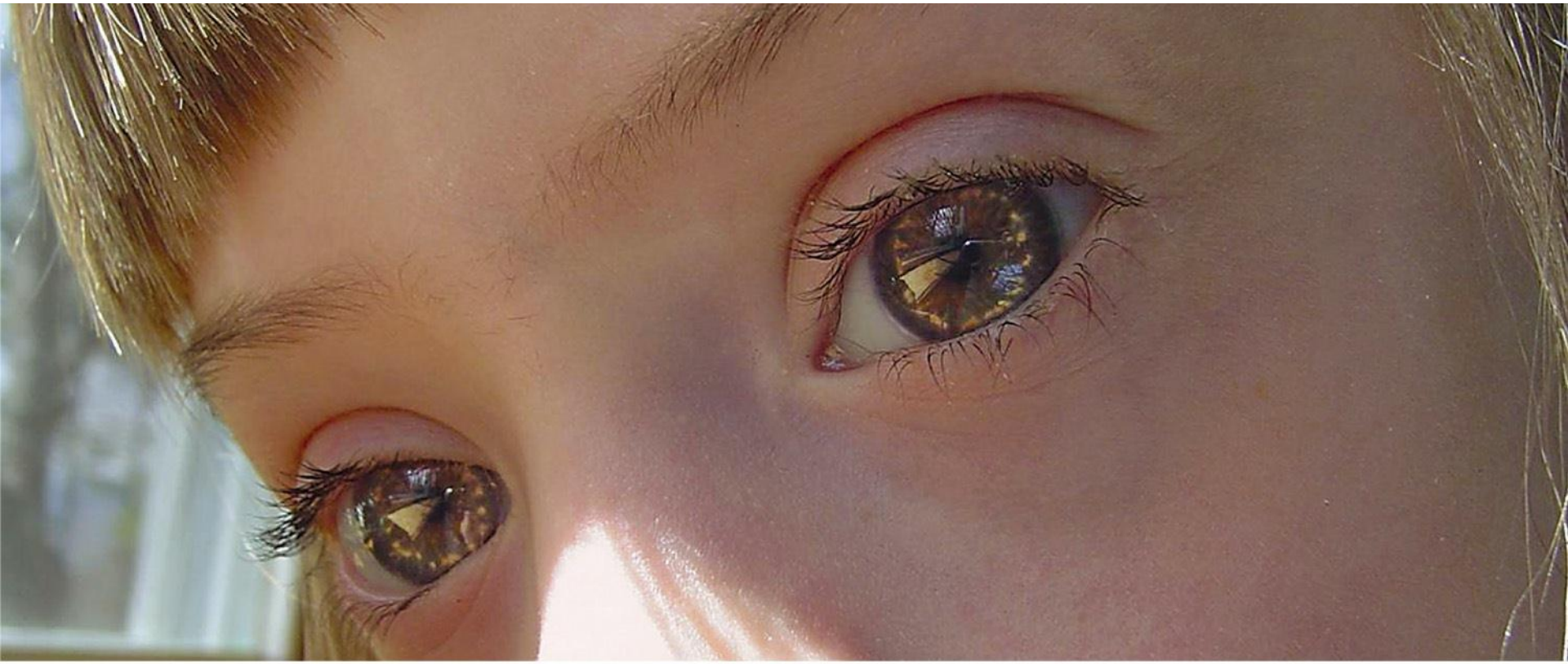
What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Aneuploidy
  - An individual's cells have too many or too few copies of a chromosome (result of nondisjunction)
  - Most cases of autosomal aneuploidy are lethal in embryos

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Trisomy 21 (Down syndrome)
  - A normal gamete (*n*) fuses with an *n*+1 gamete
  - New individual is trisomic (2*n*+1), having three of one type of chromosome and two of every other type
  - Effects: Mild to moderate mental impairment; health problems such as heart disease; flattened facial profile; fold of skin on inner corner of eye; low muscle tone
  - Occurs 1/700 births
  - Risk increases with maternal age

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)



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What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Change in number of sex chromosomes
  - Usually results in some degree of impairment in learning and motor skills
  - In individuals with trisomy (XXY, XXX, and XYY), these problems can be subtle and the cause may never be diagnosed

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Female sex chromosome abnormalities
  - Individuals with Turner syndrome have an X chromosome and no corresponding X or Y chromosome (XO)
    - Well proportioned but short
    - Ovaries do not develop properly
    - Insufficient sex hormones to become sexually mature

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

- Male sex chromosome abnormalities
  - Klinefelter syndrome (XXY)
    - Tend to be overweight
    - Tall
    - Normal range of intelligence
    - Make more estrogen and less testosterone than normal males, which has feminizing effects

What Are The Effects of Chromosome Number Changes in Humans? (cont'd.)

Disorder	Main Symptoms
Down syndrome	Mental impairment; heart defects
Turner syndrome (XO)	Sterility; abnormal ovaries and sexual traits
Klinefelter syndrome	Sterility; mild mental impairment
XXX syndrome	Minimal abnormalities
XYY condition	Mild mental impairment or no effect

14.6 How Do We Use What We Know About Human Inheritance?

- Genetic screening
  - Can estimate probability that a child will inherit a genetic disorder
  - Pedigrees and genotype are analyzed by a genetic counselor
  - Some disorders can be detected early enough to start countermeasures before symptoms develop
  - More than 30 conditions detectable prenatally

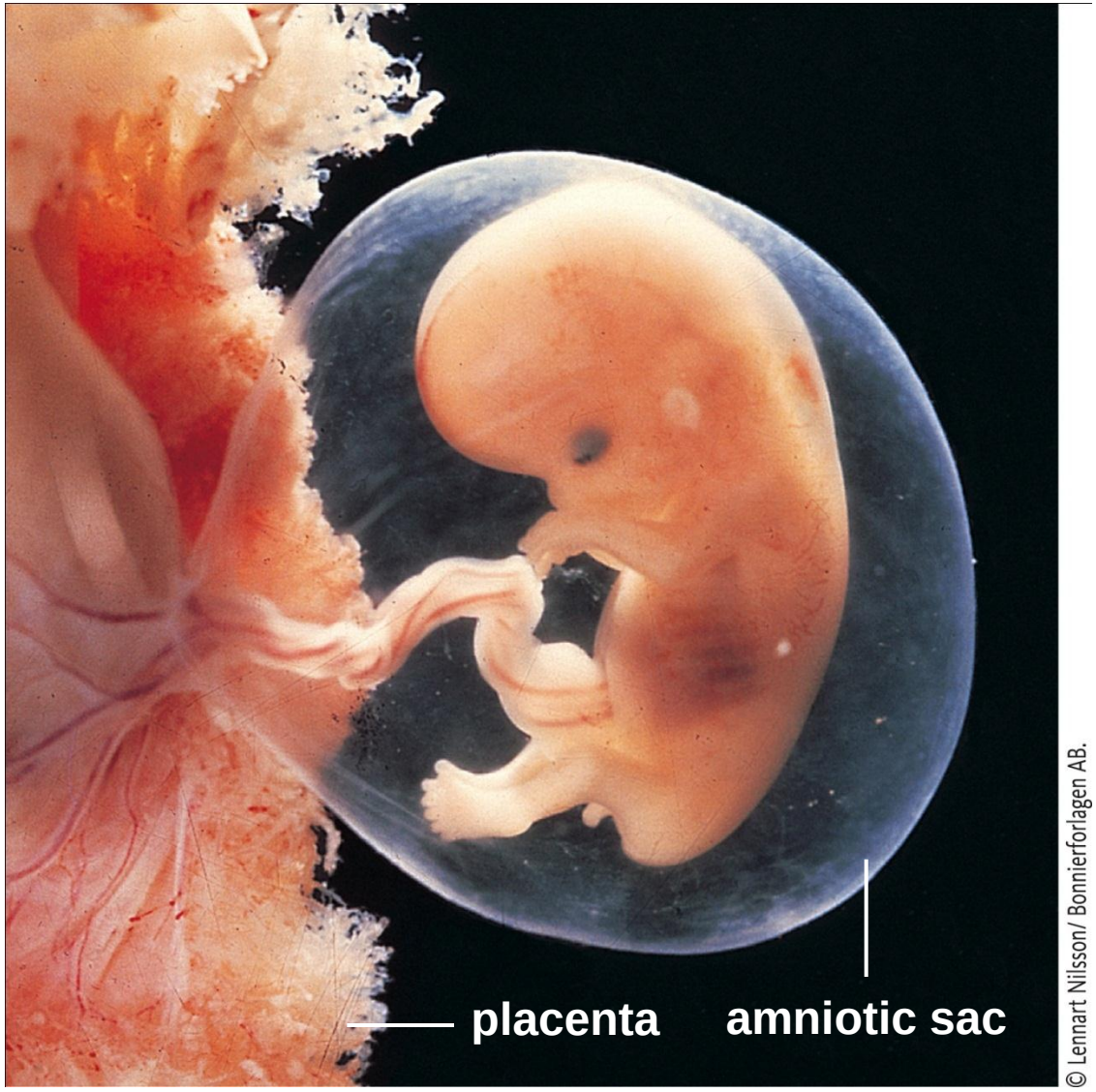
How Do We Use What We Know About Human Inheritance? (cont'd.)

- Newborn screening for phenylketonuria (PKU)
  - Newborns screened for mutations in the gene phenylalanine hydroxylase, a defect that can cause phenylalanine to accumulate to high levels
  - Results in imbalance that inhibits protein synthesis in the brain
  - Causes severe neurological symptoms characteristic of (PKU)

How Do We Use What We Know About Human Inheritance? (cont'd.)

- Prenatal diagnosis
  - Testing of an embryo or fetus can reveal genetic abnormalities or disorders before birth
    - Obstetric sonography
    - Fetoscopy
    - Amniocentesis
    - Chorionic villus sampling (CVS)
  - Invasive procedure that can carry a risk to the fetus

How Do We Use What We Know About Human Inheritance? (cont'd.)



14.7 Shades of Skin

- Skin color has a genetic basis
  - Minor differences in alleles for melanin synthesis and deposition of melanosomes affect skin color
  - Evolved as a balance between vitamin D production and protection against harmful UV radiation
  - More than 100 gene products are involved in melanin synthesis, and melanosome formation and deposition

Shades of Skin (cont'd.)

- Light-skinned people of European descent carry a mutation in gene SLC24A5 that encodes a transport protein in melanosome membranes
- People of Chinese descent carry an allele of the DCT gene that results in the conversion of tyrosine to melanin

Shades of Skin (cont'd.)

- Distribution of SLC24A5 and DCT genes suggests:
  - An African population was ancestral to both Chinese and Europeans
  - Chinese and European populations separated before their pigmentation genes mutated and their skin color changed

Shades of Skin (cont'd.)

Gary Roberts/worldwidefeatures.com.