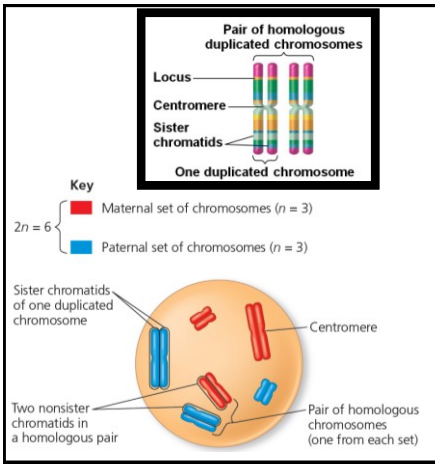
MEIOSIS AND CROSSING OVER

8.11 Chromosomes are matched in homologous pairs

- The **somatic** (body) **cells** of each species contain a specific number of chromosomes; for example, human cells have 46, consisting of 23 pairs of **homologous chromosomes**.
- The chromosomes of a homologous pair of autosomes carry genes for the same characteristics at the same place, or **locus**.
- **Checkpoint question** Do pairs of homologous chromosomes carry the same genes?

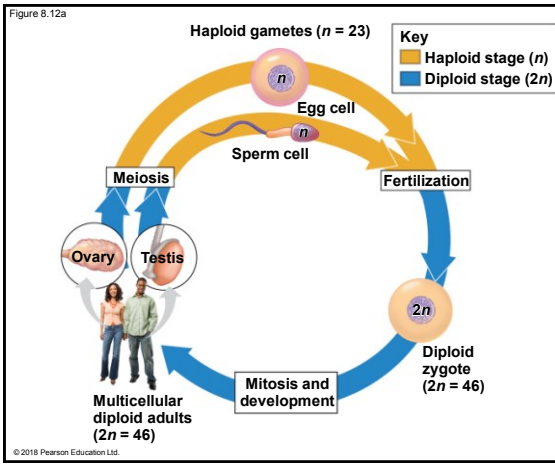


- The two chromosomes of a pair have the same length, centromere position, and staining pattern: These are called **homologous chromosomes (or homologs)**

8.12 Gametes have a single set of chromosomes

- Cells with two sets of homologous chromosomes are **diploid**.
- **Gametes**—eggs and sperm—are **haploid** cells with a single set of chromosomes.
- Sexual life cycles involve the alternation of haploid and diploid stages.

Figure 8.12a



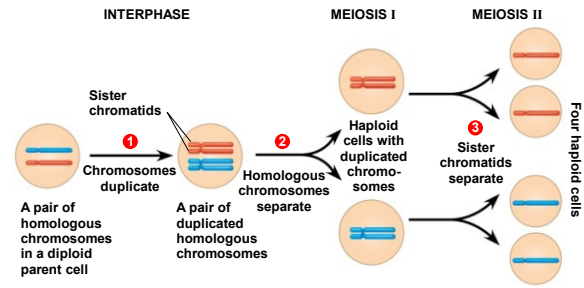
8.13 Meiosis reduces the chromosome number from diploid to haploid

- **Meiosis**, like mitosis, is preceded by chromosome duplication, but in meiosis, the cell divides twice to form four daughter cells.
  - The first division, meiosis I, starts with the pairing of homologous chromosomes.
  - In crossing over, homologous chromosomes exchange corresponding segments.

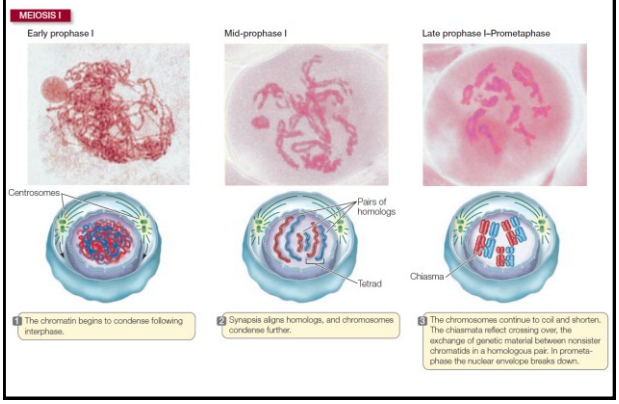
8.13 Meiosis reduces the chromosome number from diploid to haploid

- Meiosis I separates the members of each homologous pair and produces two daughter cells, each with one set of chromosomes.
- Meiosis II is essentially the same as mitosis:
  - In each of the cells, the sister chromatids of each chromosome separate.
  - The result is a total of four haploid cells.

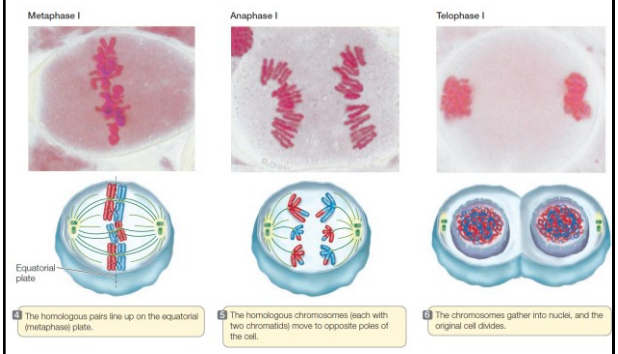
Figure 8.12b



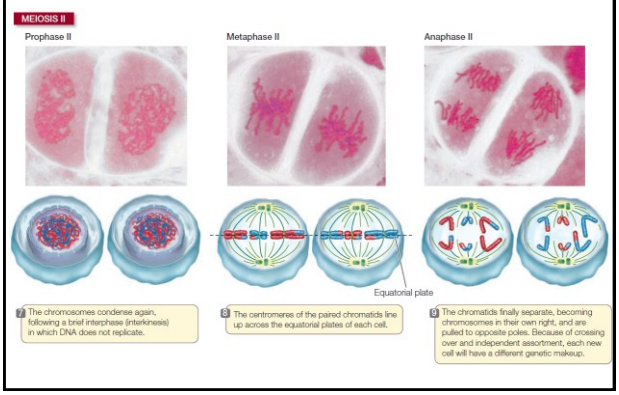
Meiosis



Meiosis (cont'd)



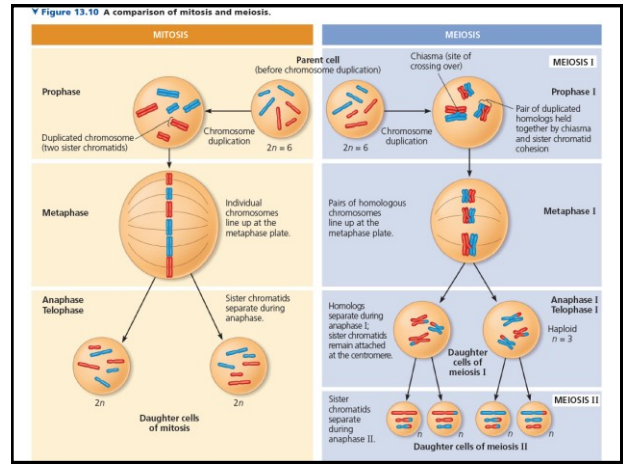
Meiosis (cont'd)



Meiosis (cont'd)

8.14 VISUALIZING THE CONCEPT: Mitosis and meiosis have important similarities and differences

- Both mitosis and meiosis begin with diploid parent cells that have chromosomes duplicated during the previous interphase.
  - Mitosis produces two genetically identical diploid somatic daughter cells.
  - Meiosis produces four genetically unique haploid gametes.

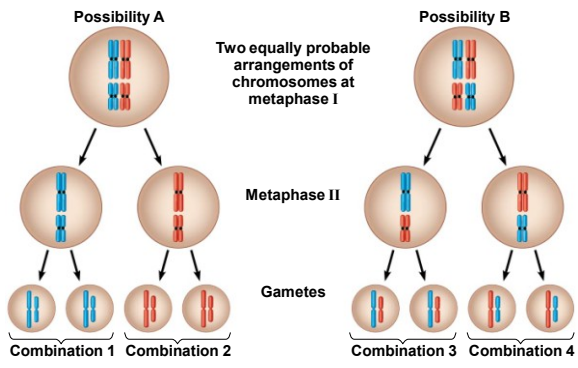


Property	Mitosis (occurs in both diploid and haploid cells)	Meiosis (can only occur in diploid cells)
DNA replication	Occurs during interphase, before mitosis begins	Occurs during interphase before meiosis I but not meiosis II
Number of divisions	One, including prophase, prometaphase, metaphase, anaphase, and telophase	Two, each including prophase, metaphase, anaphase, and telophase
Synapsis of homologous chromosomes	Does not occur	Occurs during prophase I along with crossing over between nonsister chromatids; resulting chiasmata hold pairs together due to sister chromatid cohesion
Number of daughter cells and genetic composition	Two, each genetically identical to the parent cell, with the same number of chromosomes	Four, each haploid (n); genetically different from the parent cell and from each other
Role in animals, fungi, and plants	Enables multicellular animal, fungus, or plant (gametophyte or sporophyte) to arise from a single cell; produces cells for growth, repair, and, in some species, asexual reproduction; produces gametes in the plant gametophyte	Produces gametes (in animals) or spores (in fungi and in plant sporophytes); reduces number of chromosome sets by half and introduces genetic variability among the gametes or spores

8.15 Independent orientation of chromosomes in meiosis and random fertilization lead to varied offspring

- Random arrangements of chromosome pairs at metaphase I of meiosis lead to many different combinations of chromosomes in eggs and sperm.
- Random fertilization of eggs by sperm greatly increases this variation.

Figure 8.15_3



8.16 Homologous chromosomes may carry different versions of genes

- The differences between homologous chromosomes come from the fact that they can bear different versions of genes at corresponding loci.
- **Crossing over** is an exchange of corresponding segments between nonsister chromatids of homologous chromosomes.
  - Genetic recombination, which results from crossing over during prophase I of meiosis, increases variation still further.

Figure 8.16

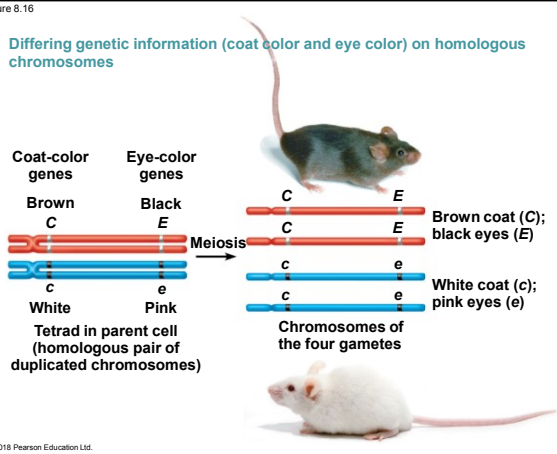
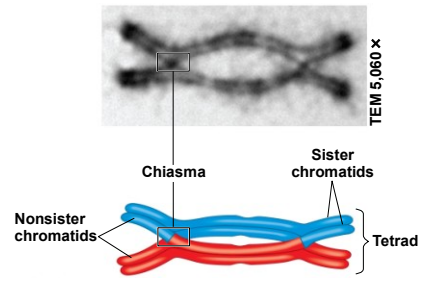


Figure 8.16b



Alterations of Chromosome Number and Structure

8.18 Accidents during meiosis can alter chromosome number

- An abnormal chromosome count can result from
  1. the failure of a pair of homologous chromosomes to separate during meiosis I or
  2. the failure of sister chromatids to separate during meiosis II.

Figure 8.18_1_3

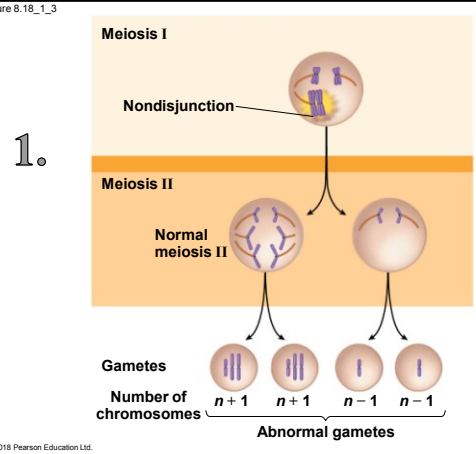
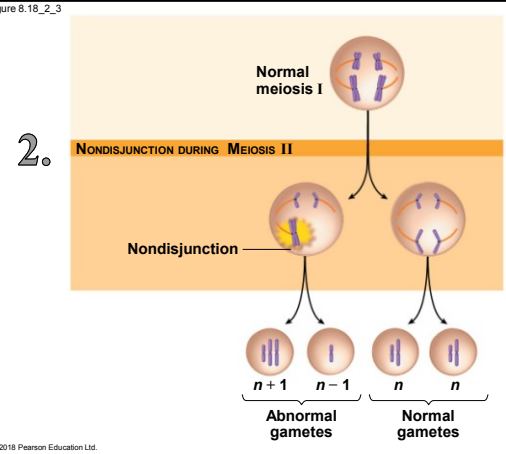


Figure 8.18_2_3



8.19 A karyotype is a photographic inventory of an individual's chromosomes

- To prepare a **karyotype**, white blood cells are
  - isolated,
  - stimulated to grow,
  - arrested at metaphase, and
  - photographed under a microscope.
- The chromosomes are arranged into ordered pairs so that any chromosomal abnormalities can be detected.

Figure 8.19_1_3

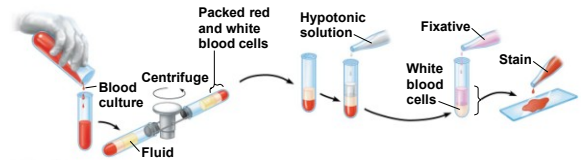


Figure 8.19_2

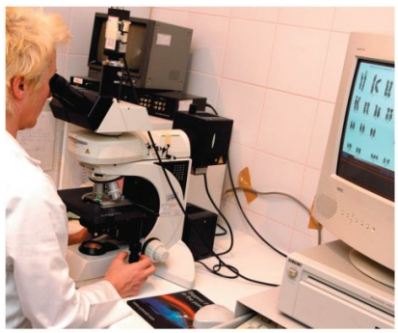
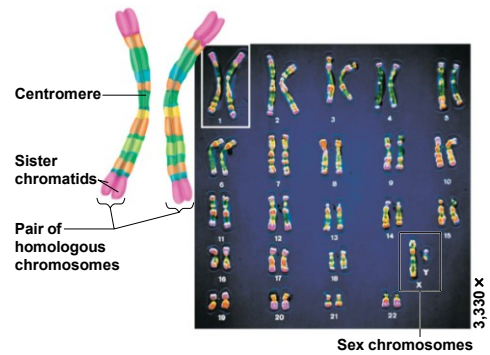
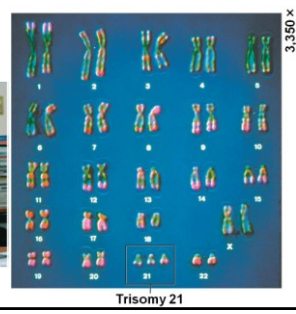


Figure 8.19_3



8.20 CONNECTION: An extra copy of chromosome 21 causes Down syndrome

- **Trisomy 21**, the most common chromosome number abnormality, results in a condition called **Down syndrome**.



8.21 CONNECTION: Abnormal numbers of sex chromosomes do not usually affect survival

- Nondisjunction of the sex chromosomes during meiosis can result in individuals with a missing or extra X or Y chromosome.
- In some cases (such as XXY), this leads to syndromes that can affect the health of the individual.
- In other cases (such as XXX), the body is normal.

TABLE 8.21 Abnormalities of Sex Chromosome Number in Humans

Sex Chromosomes	Syndrome	Origin of Nondisjunction	Symptoms
XXY	Klinefelter syndrome (male)	Meiosis in egg or sperm formation	Sterile; underdeveloped testes; secondary female characteristics
XYY	None (normal male)	Meiosis in sperm formation	None
XXX	None (normal female)	Meiosis in egg or sperm formation	Slightly taller than average
XO	Turner syndrome (female)	Meiosis in egg or sperm formation	Sterile; immature sex organs

8.22 EVOLUTION CONNECTION: New species can arise from errors in cell division

- Nondisjunction can produce polyploid organisms, organisms with extra sets of chromosomes.
- Such errors in cell division can be important in the evolution of new species.

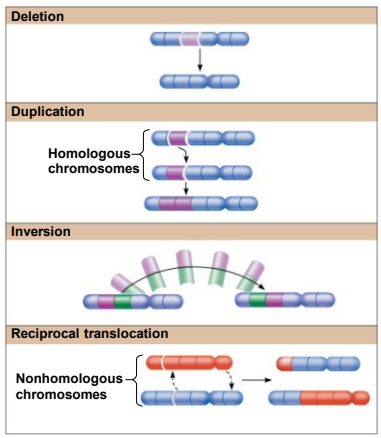
Figure 8.22 The gray tree frog (*Hyla versicolor*), a tetraploid organism

8.23 CONNECTION: Alterations of chromosome structure can cause birth defects and cancer

- Chromosome breakage can lead to rearrangements—**deletions**, **duplications**, **inversions**, and **translocations**—that can produce genetic disorders or, if the changes occur in somatic cells, cancer.

Checkpoint question How is translocation different from crossing over?

Figure 8.23a



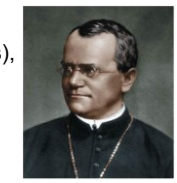
PATTERNS OF INHERITANCE

Introduction

- The Inuit people are indigenous to the arctic regions of Greenland, Canada, and Alaska.
- The traditional Inuit diet, which is high in protein and very high in fat, consists of food obtained by hunting large land mammals.
- What allows the Inuit people to tolerate high levels of dietary fat?
  - The answer lies, at least in part, in their genes.

9.2 The science of genetics began in an abbey garden

- **Heredity** is the transmission of traits from one generation to the next.
- **Genetics** (the scientific study of heredity) began with Gregor Mendel's experiments.
  - Mendel crossed pea plants and traced **traits** from generation to generation.
  - He hypothesized that there are alternative versions of genes (**alleles**), the units that determine heritable traits.



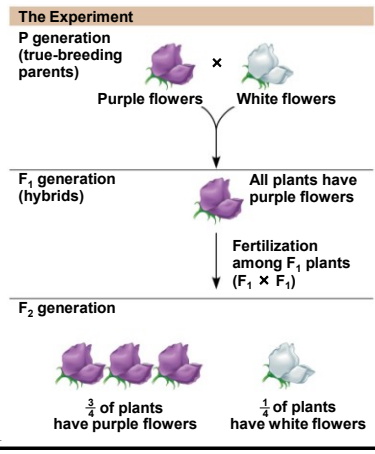
9.3 Mendel's law of segregation describes the inheritance of a single character

- Mendel developed four hypotheses, described below using modern terminology.
  1. *There are alternative versions of genes* (called **alleles**) *that account for variations in inherited characters*.
  2. *For each character, an organism inherits two alleles of a gene, one from each parent.*
     - An organism that has two identical alleles for a gene is said to be **homozygous** for that gene.
     - An organism that has two different alleles for a gene is said to be **heterozygous** for that gene.

9.3 Mendel's law of segregation describes the inheritance of a single character

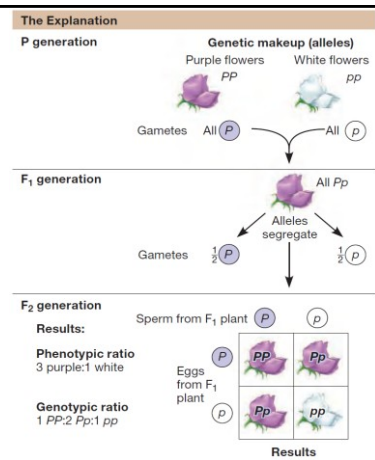
3. *If the two alleles of an inherited pair differ, then one determines the organism's appearance and is called the* **dominant allele** *and the other has no noticeable effect on the organism's appearance and is called the* **recessive allele**.
4. *A sperm or egg carries only one allele for each inherited character because allele pairs separate (segregate) from each other during the production of gametes.* This statement is called the **law of segregation**.

Figure 9.3a_3



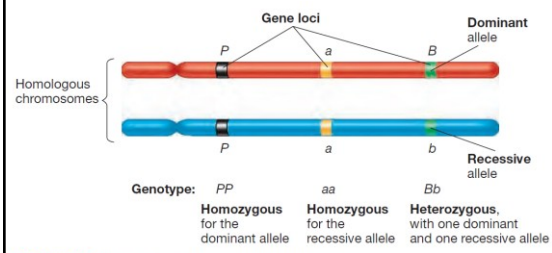
9.3 Mendel's law of segregation describes the inheritance of a single character

- Mendel's hypotheses also explain the 3:1 ratio observed in the F_2 generation.
  - The F_1 hybrids all have a *Pp* **genotype**.
  - A **Punnett square** shows the four possible combinations of alleles that could occur when these gametes combine.



9.4 Homologous chromosomes bear the alleles for each character

- Every diploid cell has pairs of homologous chromosomes.
- The chromosomes in a homologous pair carry alleles of the same genes at the same locations.



▲ **Figure 9.4** Three gene loci on homologous chromosomes

9.8 VISUALIZING THE CONCEPT: Genetic traits in humans can be tracked through family pedigrees

- The inheritance of many human traits follows Mendel's laws.
- Family **pedigrees** can help determine individual genotypes.

Figure 9.8_2

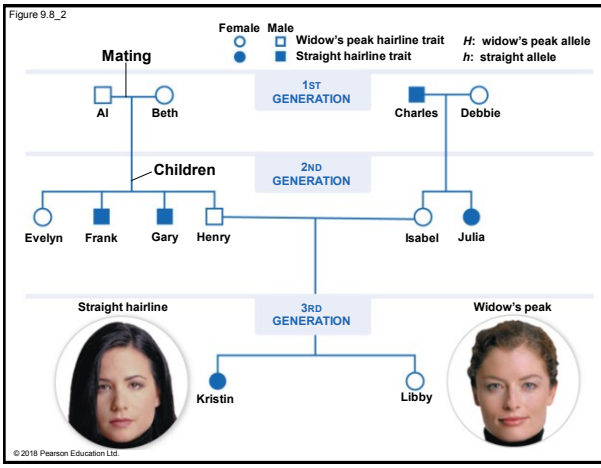


Figure 9.8_3

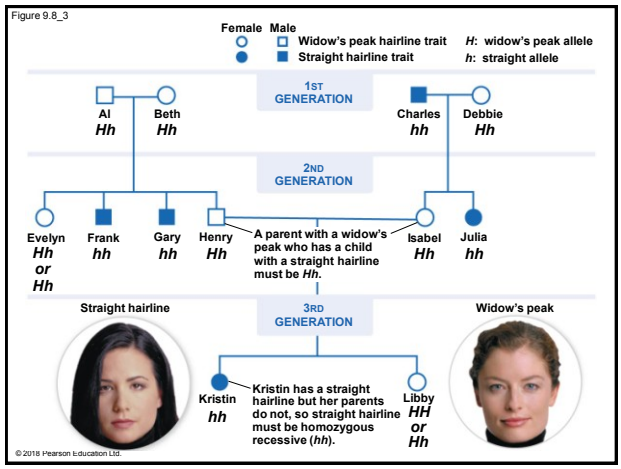
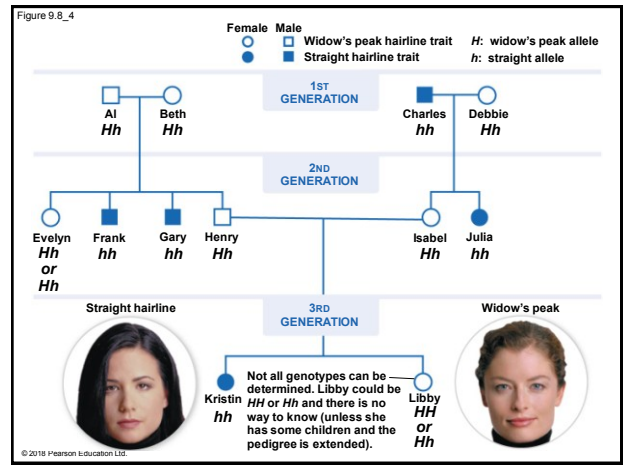


Figure 9.8_4



9.9 CONNECTION: Many inherited traits in humans are controlled by a single gene

- Most people who have recessive disorders are born to normal parents who are both heterozygotes—that is, parents who are **carriers** of the recessive allele for the disorder but are phenotypically normal.

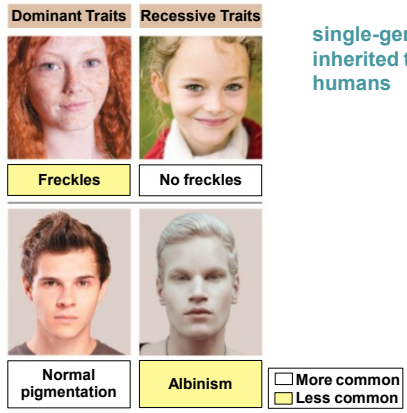
Table 9.9

The genetic disorders below are known to be inherited as dominant or recessive traits controlled by a single gene.

TABLE 9.9 Some Autosomal Disorders in Humans

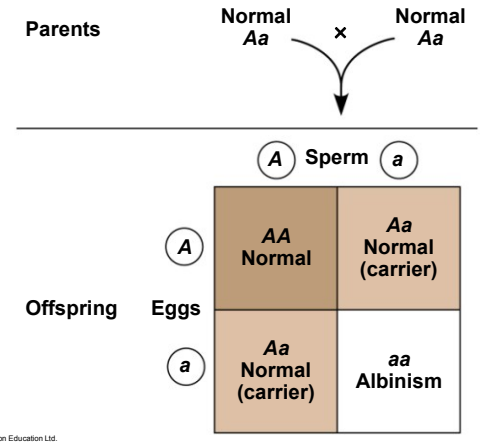
Disorder	Major Symptoms
Recessive Disorders	
Albinism	Lack of pigment in the skin, hair, and eyes
Cystic fibrosis	Excess mucus in the lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; developmental disabilities
Sickle-cell disease	Sickled red blood cells; damage to many tissues
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood
Dominant Disorders	
Achondroplasia	Dwarfism
Huntington's disease	Developmental disabilities and uncontrollable movements; cognitive impairments; strikes in middle age
Hypercholesterolemia	Excess cholesterol in the blood; heart disease

Figure 9.9a



single-gene inherited traits in humans

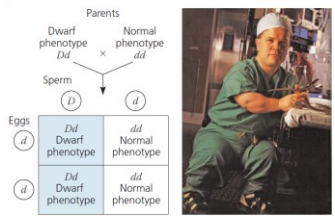
Figure 9.9b



Dominantly inherited disorders

- Although many harmful alleles are recessive, a number of human disorders are due to dominant alleles.
- **Achondroplasia**, a form of dwarfism that occurs in one of every 25,000 people. Heterozygous individuals have the dwarf phenotype. Therefore, all people who do not have achondroplasia—99.99% of the population—are homozygous for the recessive allele.

Figure 14.18 Achondroplasia: a dominant trait.
Dr. Michael C. Ain has achondroplasia, a form of dwarfism caused by a dominant allele. This has inspired his work: He is a specialist in the repair of bone defects caused by achondroplasia and other disorders. The dominant allele (*D*) might have arisen as a mutation in the egg or sperm of a parent or could have been inherited from an affected parent, as shown for an affected father in the Punnett square.



VARIATIONS ON MENDEL'S LAWS

9.12 Many genes have more than two alleles that may be codominant

- The **ABO blood group** phenotype in humans is controlled by three alleles that produce a total of four phenotypes.
- The I^A and I^B alleles are **codominant**: Both alleles are expressed in heterozygous individuals (I^AI^B), who have type AB blood.

9.12 Many genes have more than two alleles that may be codominant

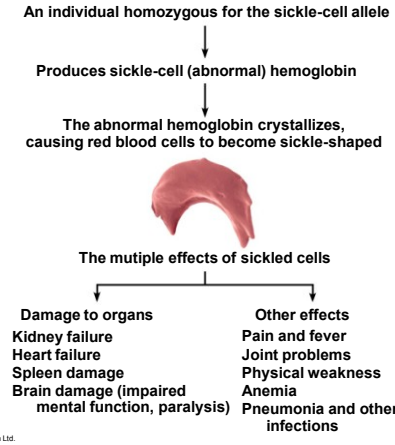
- **Checkpoint question** Steven has type B blood, and Lucy has type A blood. Can Aaron, who has type O blood, be their biological child?

Blood Group (Phenotype)	Genotypes	Carbohydrates Present on Red Blood Cells
A	I^AI^A or I^Ai	Carbohydrate A
B	I^BI^B or I^Bi	Carbohydrate B
AB	I^AI^B	Carbohydrate A and Carbohydrate B
O	ii	Neither

9.13 A single gene may affect many phenotypic characters

- **Pleiotropy** occurs when one gene influences multiple characters.
- **Sickle-cell disease** is a human example of pleiotropy.
  - This disease affects the type of hemoglobin produced and the shape of red blood cells, and causes anemia and organ damage.
  - Sickle-cell and nonsickle alleles are codominant.
  - Carriers of sickle-cell disease have increased resistance to malaria.

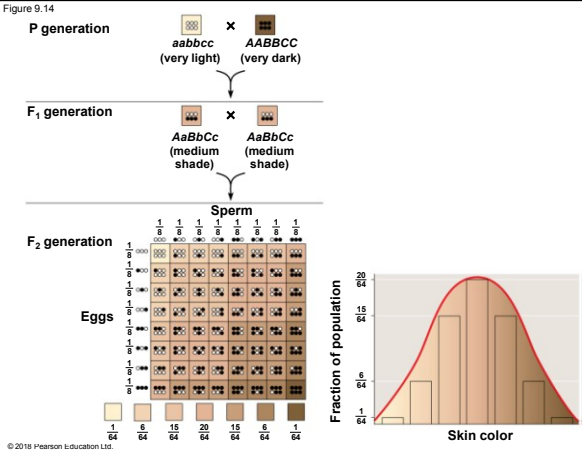
Figure 9.13b



9.14 A single character may be influenced by many genes

- Many characters result from **polygenic inheritance**, in which a single phenotypic character results from the additive effects of two or more genes on a single phenotypic character.
- Human skin color is an example of polygenic inheritance.

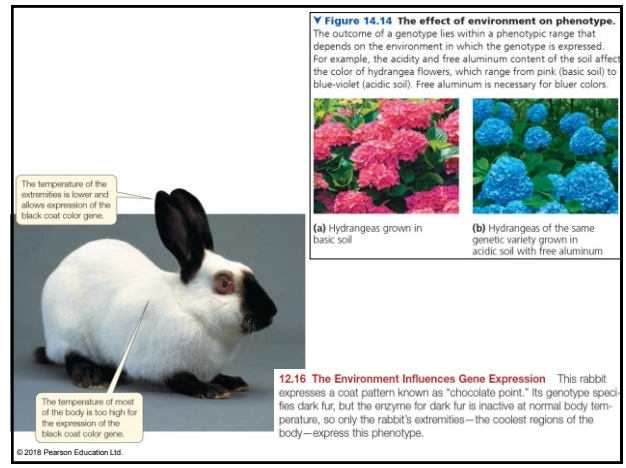
Figure 9.14



9.15 The environment affects many characters

- Many traits are affected, in varying degrees, by both genetic and environmental factors.

	Rare	Common
	Simple genetics	Complex genetics
	High recurrence risk	Low recurrence risk
← genetic		environmental →
Sickle cell disease	Hypertension Heart disease Diabetes Asthma Behavioral disorders	Infectious diseases

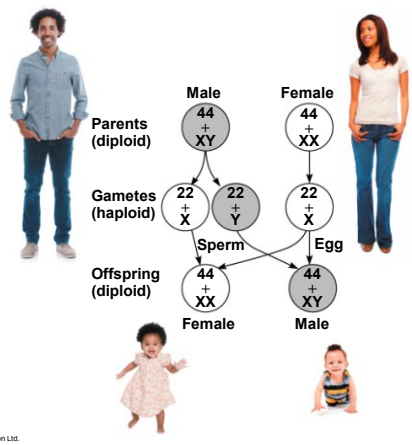


Sex Chromosomes and Sex-Linked Genes

9.20 Chromosomes determine sex in many species

- In mammals, a male has XY **sex chromosomes**, and a female has XX.
  - The Y chromosome has genes for the development of testes, whereas an absence of the Y allows ovaries to develop.
  - In addition, human males and females both have 44 autosomes (nonsex chromosomes).
- Other systems of sex determination exist in other animals and plants.

Figure 9.20b



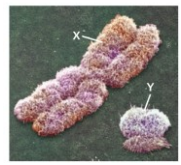
9.20 Chromosomes determine sex in many species

- In some animals, environmental temperature determines the sex.
  - For some reptiles, the temperature at which eggs are incubated during a specific period of embryonic development determines whether that embryo will develop into a male or female.
  - Global climate change may therefore impact the sex ratio of such species.

9.21 Sex-linked genes exhibit a unique pattern of inheritance

- A gene located on either sex chromosome is called a **sex-linked gene**.
- Y chromosome contains the maleness-determining gene, which was named *SRY (sex-determining region on the Y chromosome).*
- The *SRY* gene encodes a protein involved in **primary sex determination**
- In the presence of the functional SRY protein, an embryo develops sperm-producing testes
- In the absence of *SRY,* ovaries develop, even in an XY embryo.
  - Some women are genetically XY but lack a small portion of the Y chromosome.
  - Some men are genetically XX but have a small piece of the Y chromosome attached to another chromosome.

Y Figure 15.5 Human sex chromosomes.



9.21 Sex-linked genes exhibit a unique pattern of inheritance

- The X chromosome carries many **X-linked genes** that control traits unrelated to sex.
- The inheritance of white eye color in the fruit fly illustrates an X-linked recessive trait.

Figure 9.21b

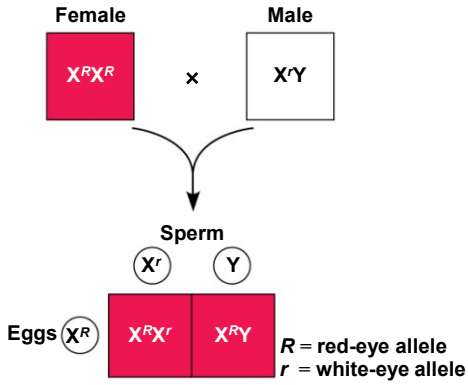


Figure 9.21c

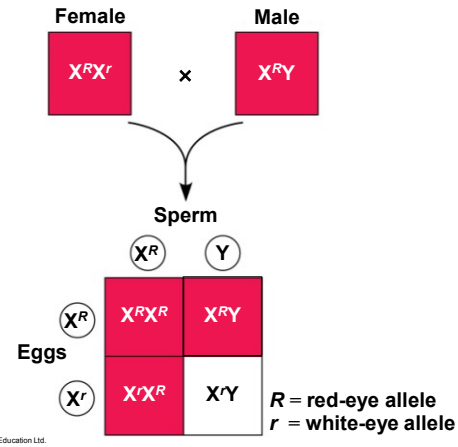
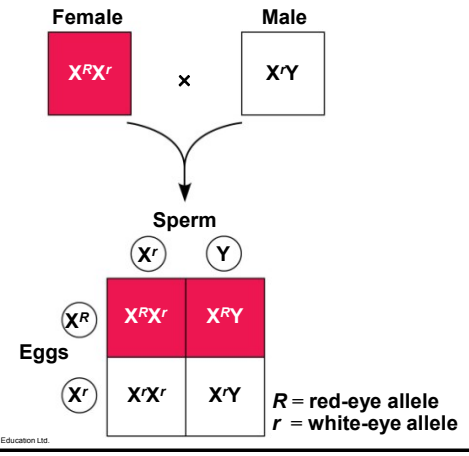
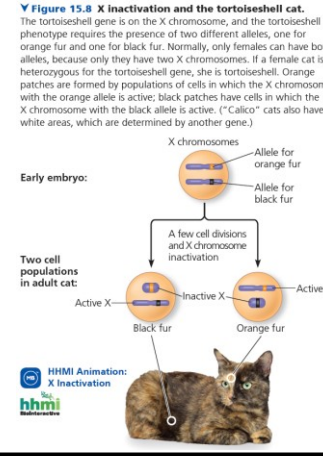


Figure 9.21d



- Almost all of one X chromosome in each cell in female mammals becomes inactivated during early embryonic development. As a result, the cells of females and males have only one active copy of most X-linked genes. The inactive X in each cell of a female condenses into a compact object called a **Barr body (discovered by Canadian anatomist Murray Barr)** Most of the genes of the X chromosome that forms the Barr body are not expressed.

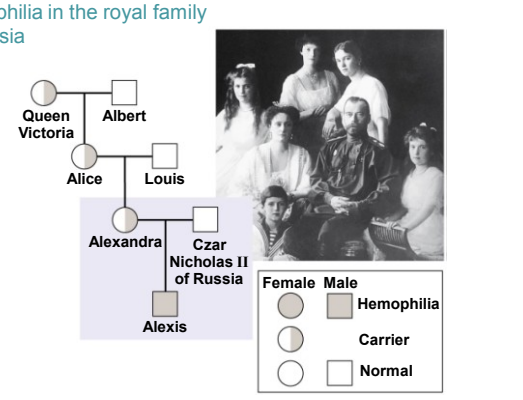


9.22 CONNECTION: Human sex-linked disorders affect mostly males

- Most X-linked human disorders are due to recessive alleles and therefore are seen mostly in males.
  - A male receiving a single X-linked recessive allele from his mother will have the disorder.
  - A female must receive the allele from both parents to be affected.

Figure 9.22

Hemophilia in the royal family of Russia



9.23 EVOLUTION CONNECTION: The Y chromosome provides clues about human male evolution

- Y chromosomes can provide data about recent human evolutionary history because they are passed on intact from father to son.
- Like the Y chromosome, mitochondrial DNA (mtDNA) can be used to trace maternal ancestry because mitochondria are characteristically inherited from the egg.

9.23 EVOLUTION CONNECTION: The Y chromosome provides clues about human male evolution

- In 2003, geneticists discovered that about 8% of males currently living in central Asia have Y chromosomes of striking genetic similarity.
  - Further analysis traced their common genetic heritage to a man living about 1,000 years ago.
  - In combination with historical records, the data led to the speculation that the Mongolian ruler Genghis Khan may be responsible for the spread of the telltale chromosome to nearly 16 million male descendants.

9.10 CONNECTION: New technologies can provide insight into one's genetic legacy

- Carrier screening, fetal testing, fetal imaging, and newborn screening can provide information for reproductive decisions but may create ethical dilemmas.

Checkpoint question What is the primary benefit of genetic screening by CVS? What is the primary risk?

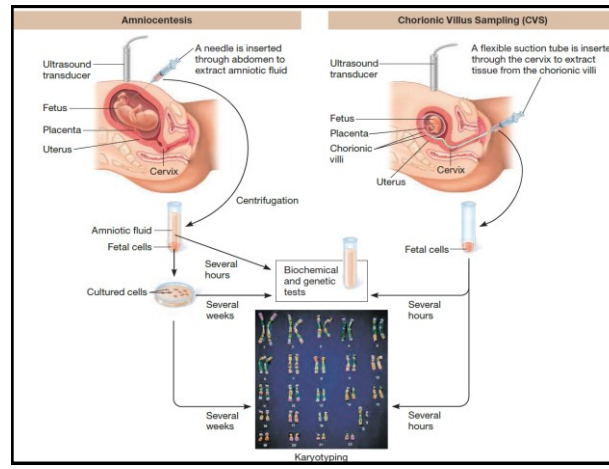
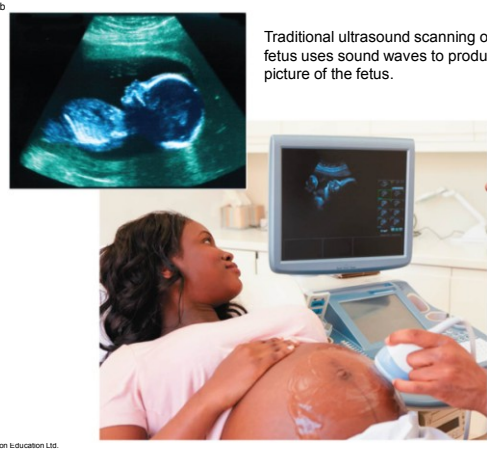


Figure 9.10b



Traditional ultrasound scanning of a fetus uses sound waves to produce a picture of the fetus.

Video: Ultrasound of Human Fetus