Overview: Variations on a Theme

- Living organisms are distinguished by their ability to reproduce their own kind
- **Genetics** is the scientific study of heredity and variation
- **Heredity** is the transmission of traits from one generation to the next
- **Variation** is demonstrated by the differences in appearance that offspring show from parents and siblings

Figure 13.1

1: Offspring acquire genes from parents by inheriting chromosomes

- In a literal sense, children do not inherit particular physical traits from their parents
- It is genes that are actually inherited

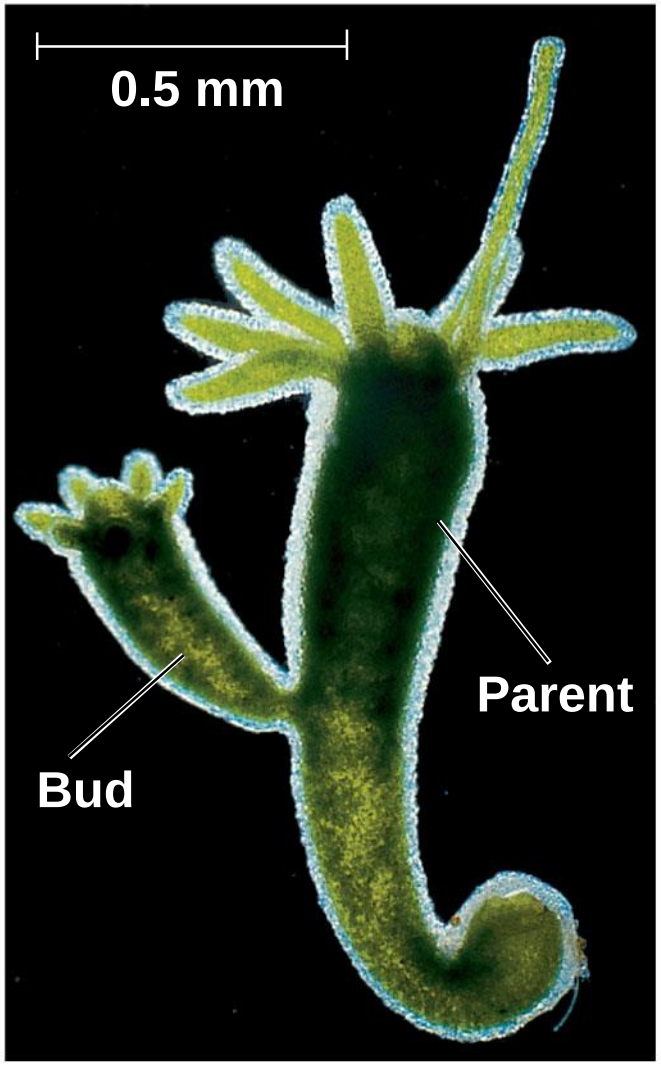
Inheritance of Genes

- **Genes** are the units of heredity, and are made up of segments of DNA
- Genes are passed to the next generation via reproductive cells called **gametes** (sperm and eggs)
- Each gene has a specific location called a **locus** on a certain chromosome
- Most DNA is packaged into chromosomes

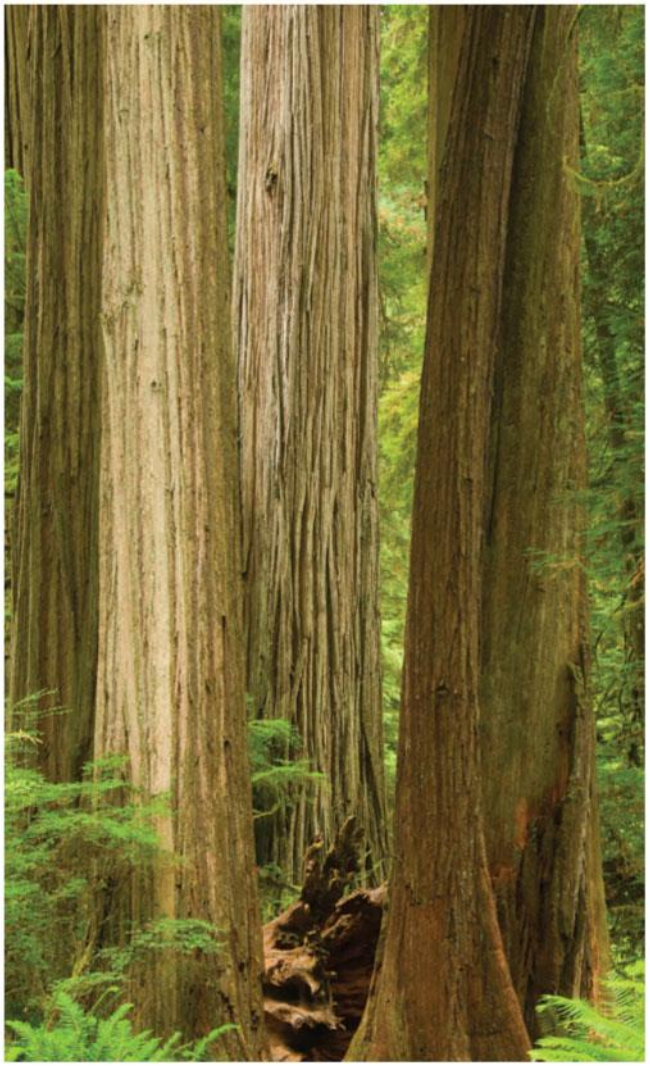
Comparison of Asexual and Sexual Reproduction

- In **asexual reproduction**, a single individual passes genes to its offspring without the fusion of gametes
- A **clone** is a group of genetically identical individuals from the same parent
- In **sexual reproduction**, two parents give rise to offspring that have unique combinations of genes inherited from the two parents

Figure 13.2



(a) Hydra



(b) Redwoods

2: Fertilization and meiosis alternate in sexual life cycles

- A **life cycle** is the generation-to-generation sequence of stages in the reproductive history of an organism

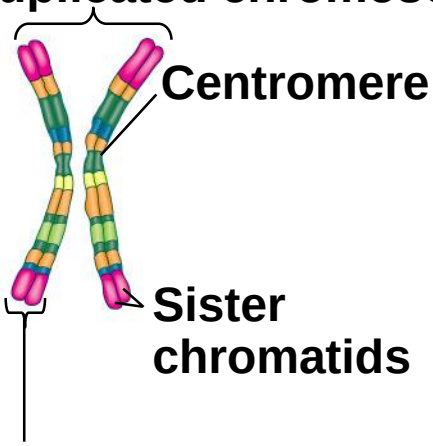
Sets of Chromosomes in Human Cells

- Human **somatic cells** (any cell other than a gamete) have 23 pairs of chromosomes
- A **karyotype** is an ordered display of the pairs of chromosomes from a cell
- The two chromosomes in each pair are called **homologous chromosomes**, or homologs
- Chromosomes in a homologous pair are the same length and shape and carry genes controlling the same inherited characters

APPLICATION

TECHNIQUE

Pair of homologous duplicated chromosomes



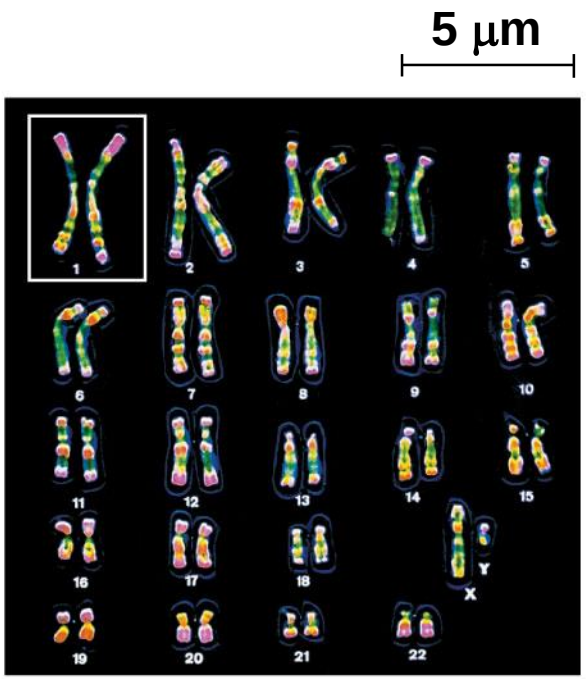
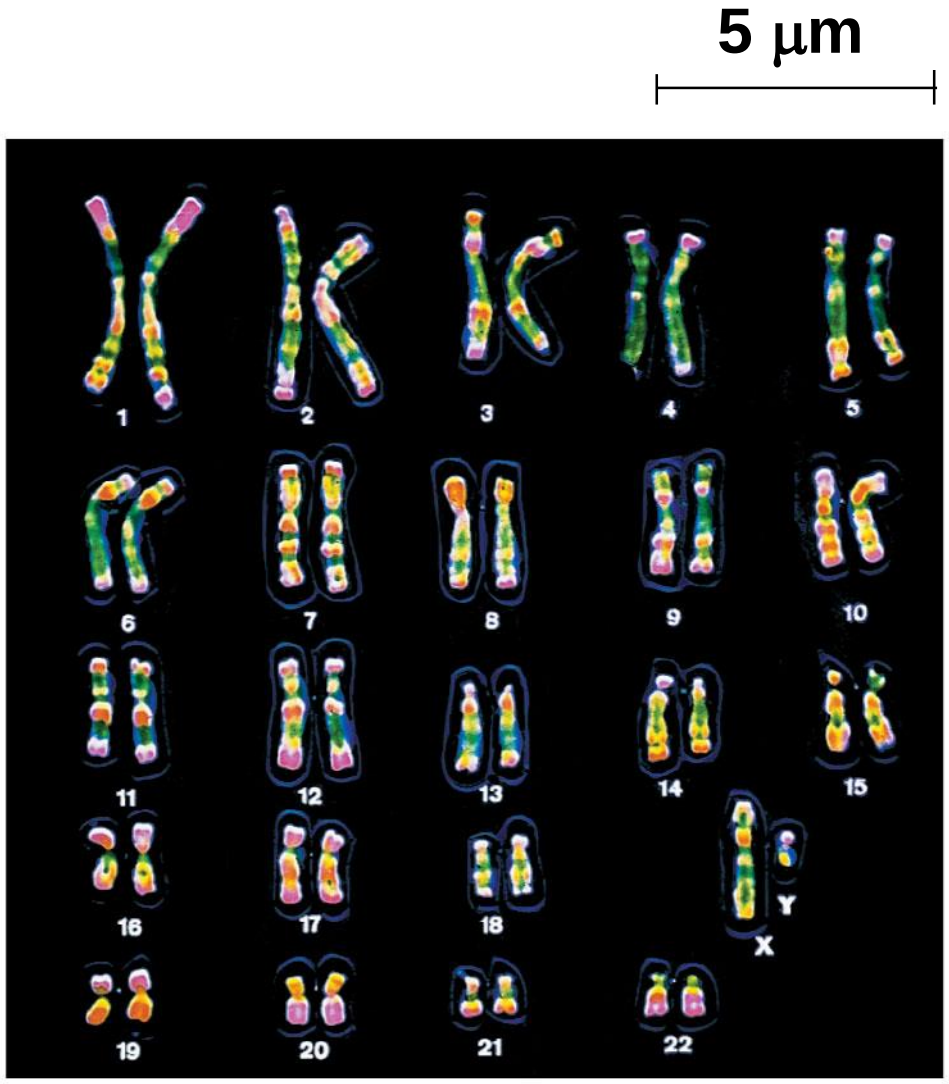


Figure 13.3c

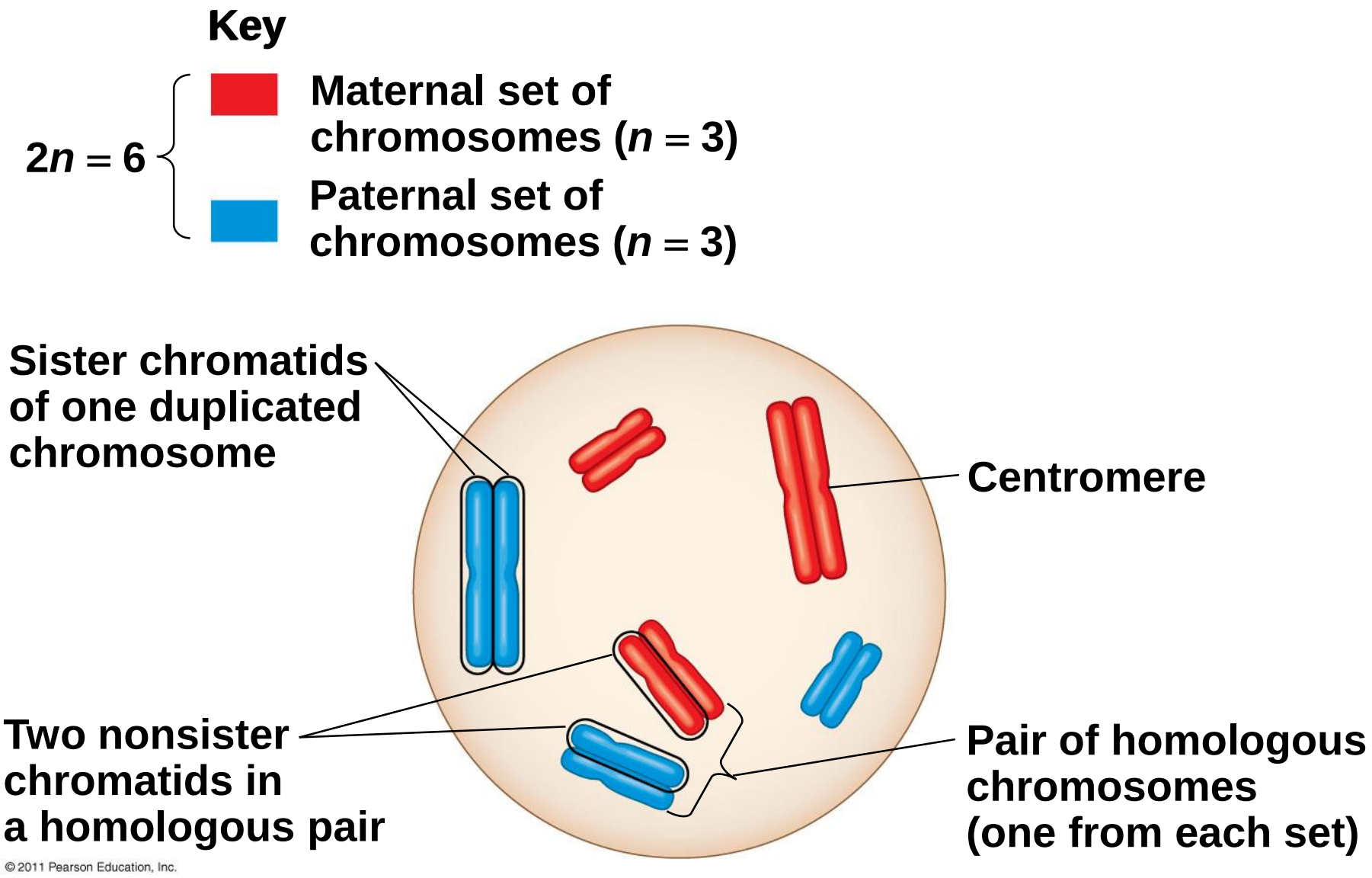


- The **sex chromosomes**, which determine the sex of the individual, are called X and Y
- Human females have a homologous pair of X chromosomes (XX)
- Human males have one X and one Y chromosome
- The remaining 22 pairs of chromosomes are called **autosomes**

- Each pair of homologous chromosomes includes one chromosome from each parent
- The 46 chromosomes in a human somatic cell are two sets of 23: one from the mother and one from the father
- A **diploid cell** (2*n*) has two sets of chromosomes
- For humans, the diploid number is 46 (2*n* = 46)

- In a cell in which DNA synthesis has occurred, each chromosome is replicated
- Each replicated chromosome consists of two identical sister chromatids

Figure 13.4

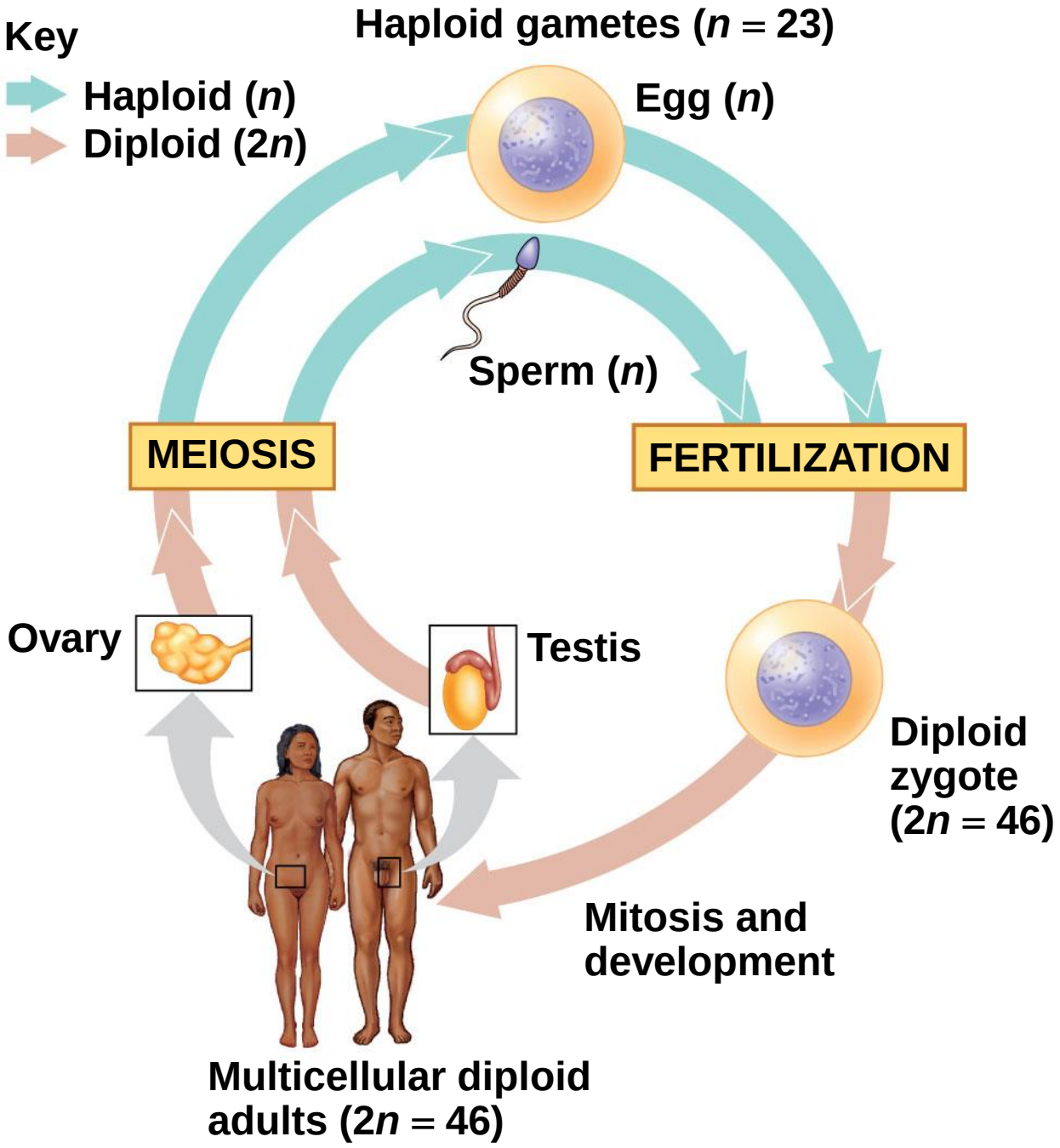


- A gamete (sperm or egg) contains a single set of chromosomes, and is **haploid** (*n*)
- For humans, the haploid number is 23 (*n* = 23)
- Each set of 23 consists of 22 autosomes and a single sex chromosome
- In an unfertilized egg (ovum), the sex chromosome is X
- In a sperm cell, the sex chromosome may be either X or Y

Behavior of Chromosome Sets in the Human Life Cycle

- **Fertilization** is the union of gametes (the sperm and the egg)
- The fertilized egg is called a **zygote** and has one set of chromosomes from each parent
- The zygote produces somatic cells by mitosis and develops into an adult

- At sexual maturity, the ovaries and testes produce haploid gametes
- Gametes are the only types of human cells produced by **meiosis**, rather than mitosis
- Meiosis results in one set of chromosomes in each gamete
- Fertilization and meiosis alternate in sexual life cycles to maintain chromosome number

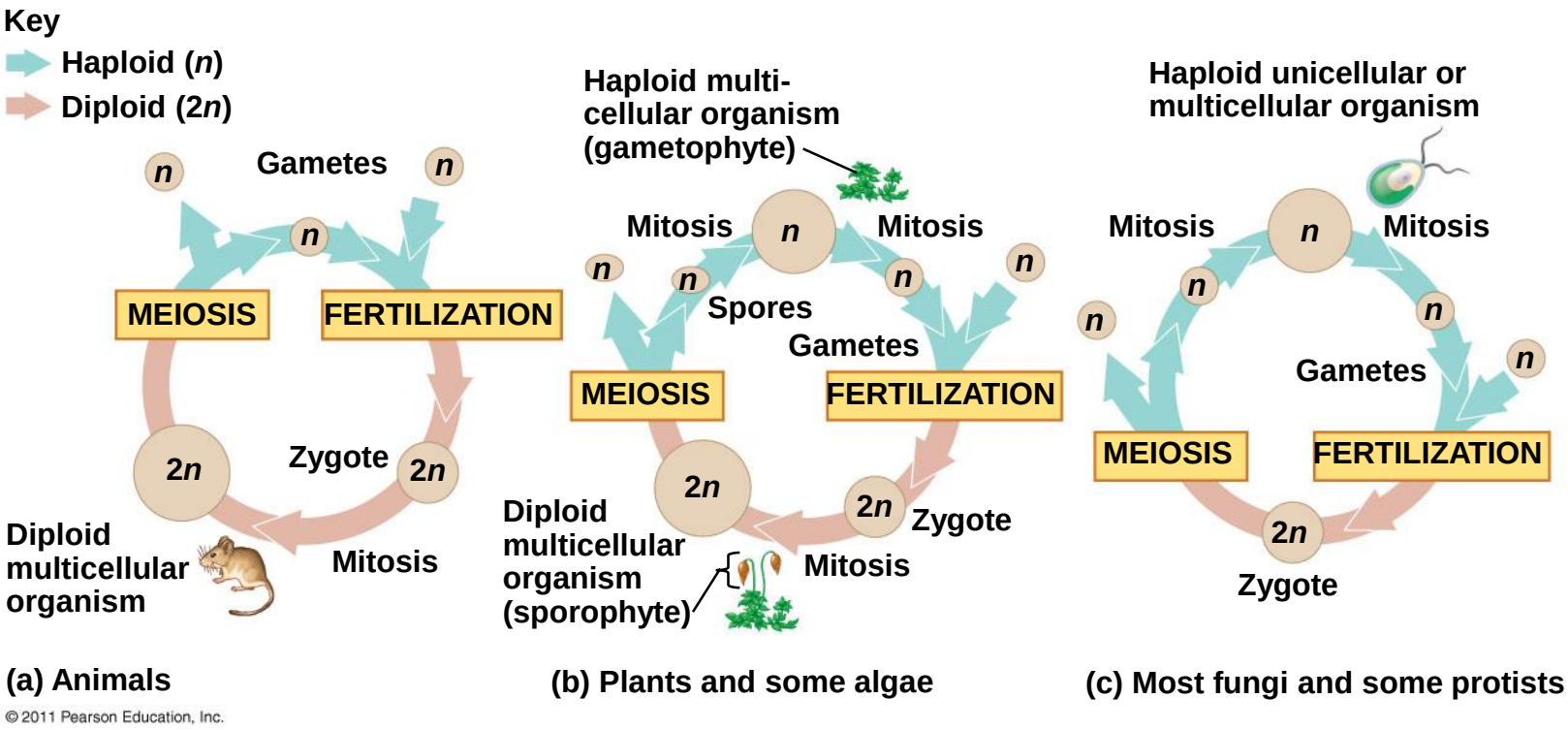


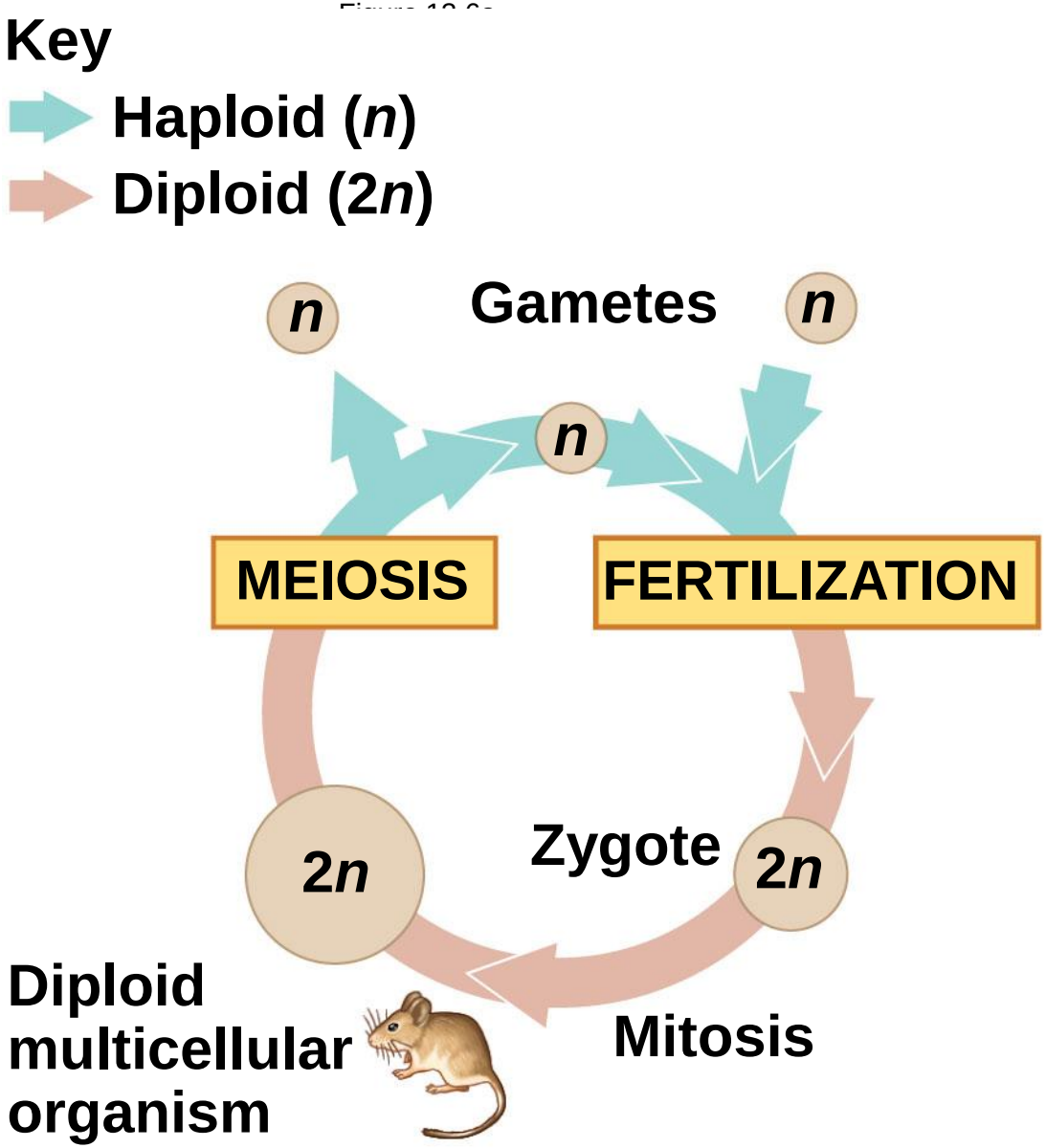
The Variety of Sexual Life Cycles

- The alternation of meiosis and fertilization is common to all organisms that reproduce sexually
- The three main types of sexual life cycles differ in the timing of meiosis and fertilization

- Gametes are the only haploid cells in animals
- They are produces by meiosis and undergo no further cell division before fertilization
- Gametes fuse to form a diploid zygote that divides by mitosis to develop into a multicellular organism

Figure 13.6

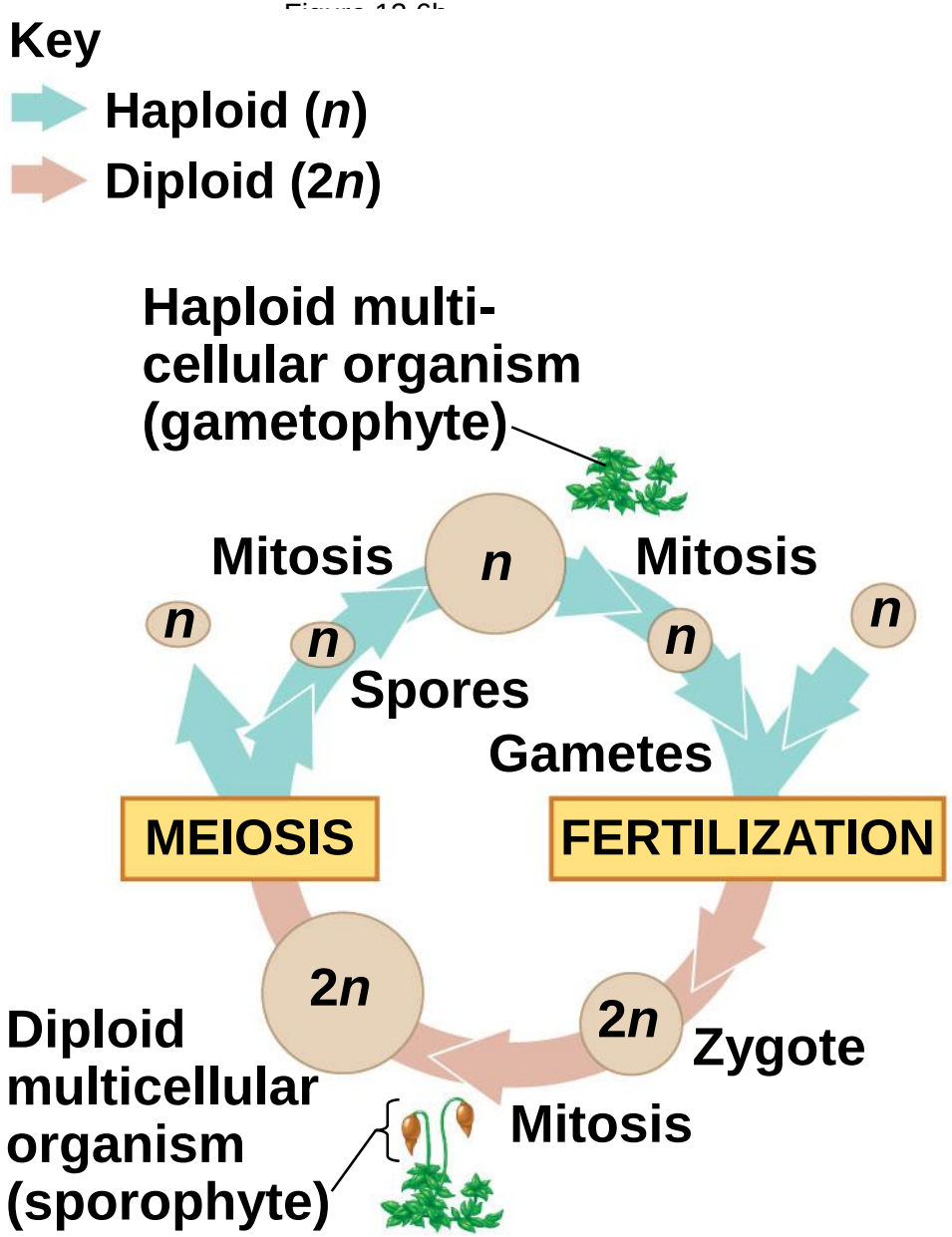




(a) Animals

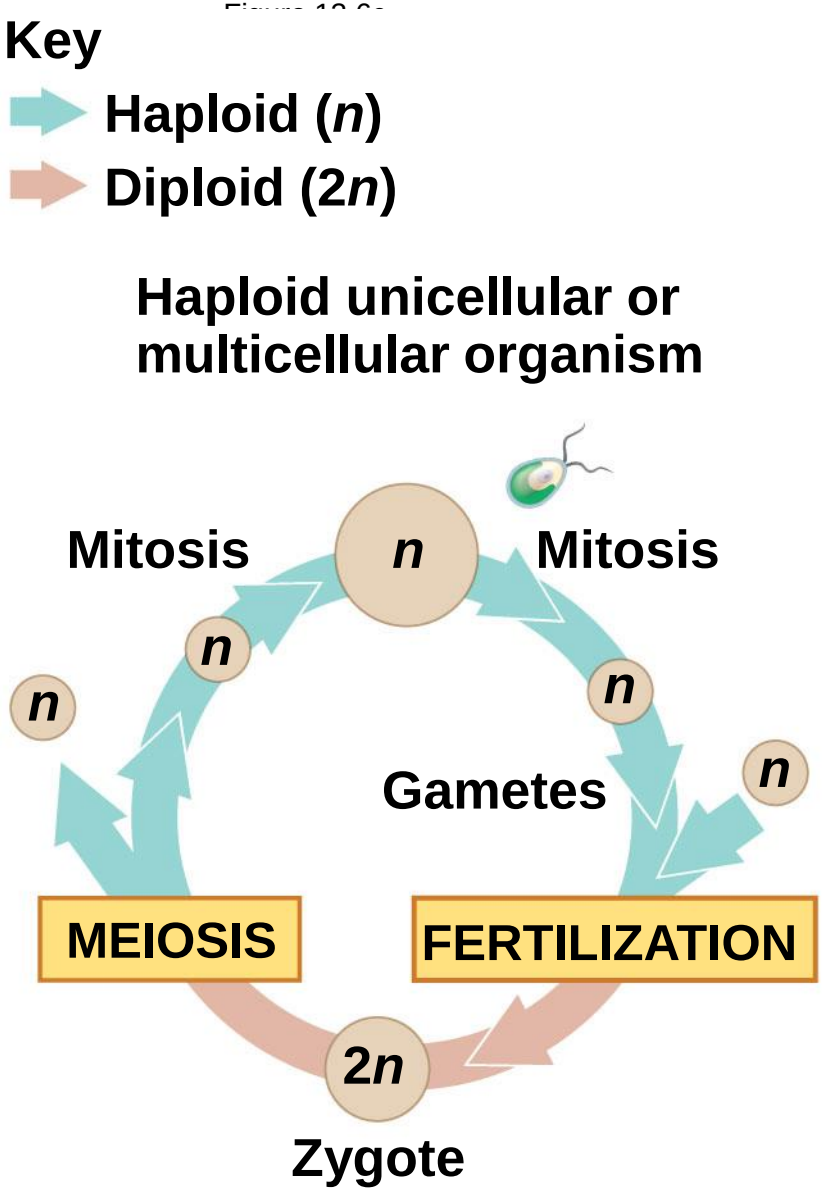
- Plants and some algae exhibit an **alternation of generations**
- This life cycle includes both a diploid and haploid multicellular stage
- The diploid organism, called the sporophyte, makes haploid spores by meiosis

- Each spore grows by mitosis into a haploid organism called a gametophyte
- A gametophyte makes haploid gametes by mitosis
- Fertilization of gametes results in a diploid sporophyte



(b) Plants and some algae

- In most fungi and some protists, the only diploid stage is the single-celled zygote; there is no multicellular diploid stage
- The zygote produces haploid cells by meiosis
- Each haploid cell grows by mitosis into a haploid multicellular organism
- The haploid adult produces gametes by mitosis



(c) Most fungi and some protists

- Depending on the type of life cycle, either haploid or diploid cells can divide by mitosis
- However, only diploid cells can undergo meiosis
- In all three life cycles, the halving and doubling of chromosomes contributes to genetic variation in offspring

3: Meiosis reduces the number of chromosome sets from diploid to haploid

- Like mitosis, meiosis is preceded by the replication of chromosomes
- Meiosis takes place in two sets of cell divisions, called **meiosis I** and **meiosis II**
- The two cell divisions result in four daughter cells, rather than the two daughter cells in mitosis
- Each daughter cell has only half as many chromosomes as the parent cell

The Stages of Meiosis

- After chromosomes duplicate, two divisions follow
  - Meiosis I (reductional division): homologs pair up and separate, resulting in two haploid daughter cells with replicated chromosomes
  - Meiosis II (equational division) sister chromatids separate
- The result is four haploid daughter cells with unreplicated chromosomes

- Meiosis I is preceded by interphase, when the chromosomes are duplicated to form sister chromatids
- The sister chromatids are genetically identical and joined at the centromere
- The single centrosome replicates, forming two centrosomes

- Division in meiosis I occurs in four phases
  - Prophase I
  - Metaphase I
  - Anaphase I
  - Telophase I and cytokinesis

Figure 13.8

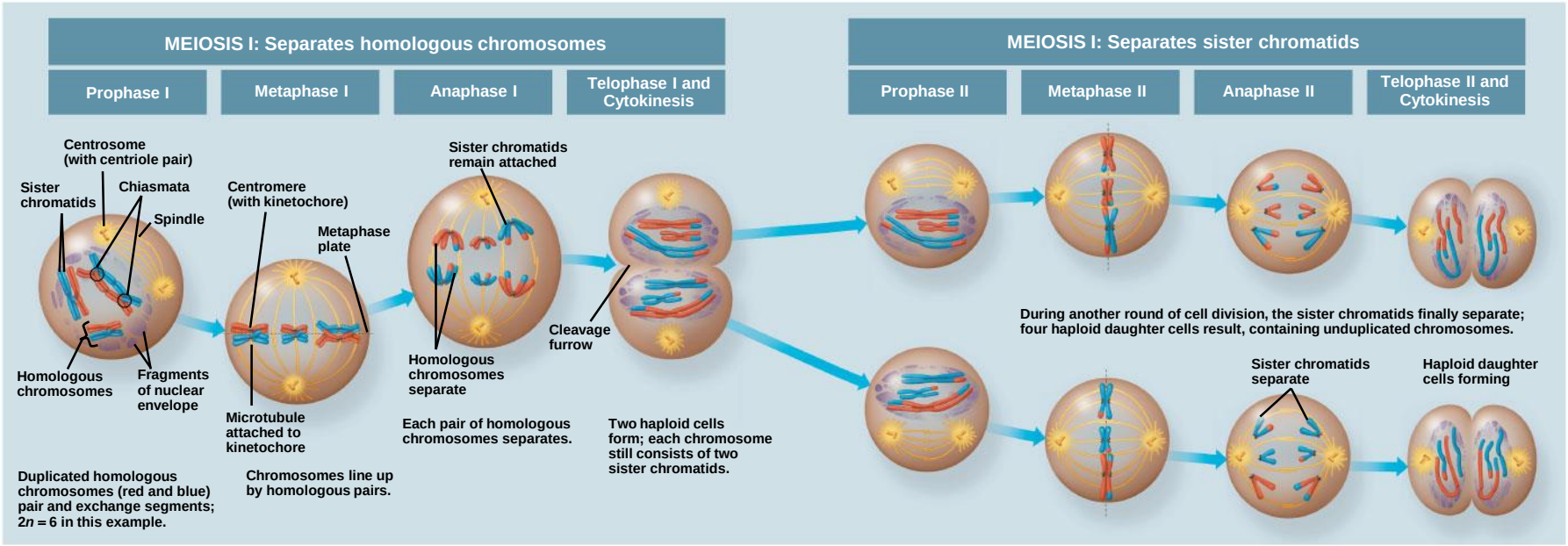
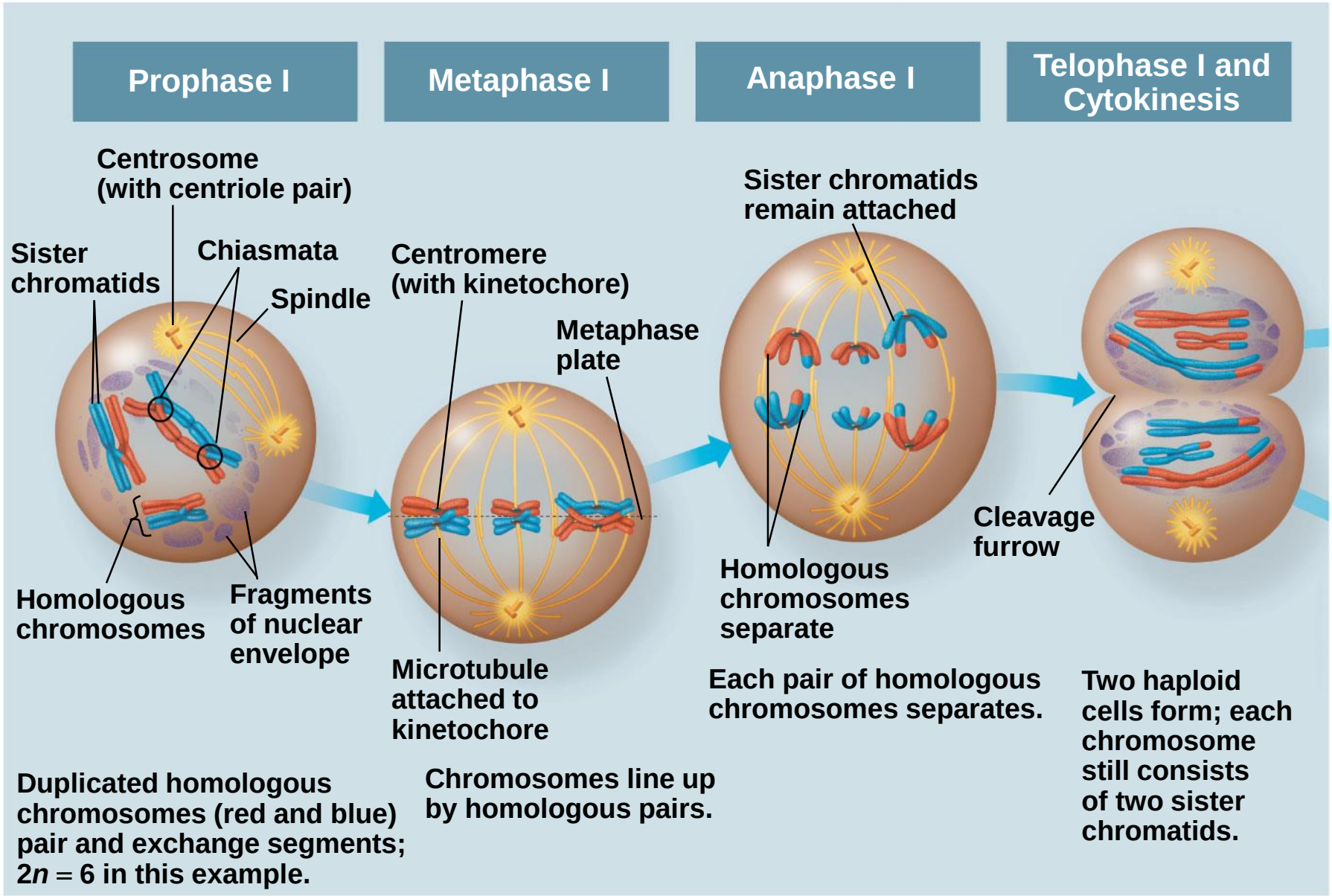


Figure 13.8a



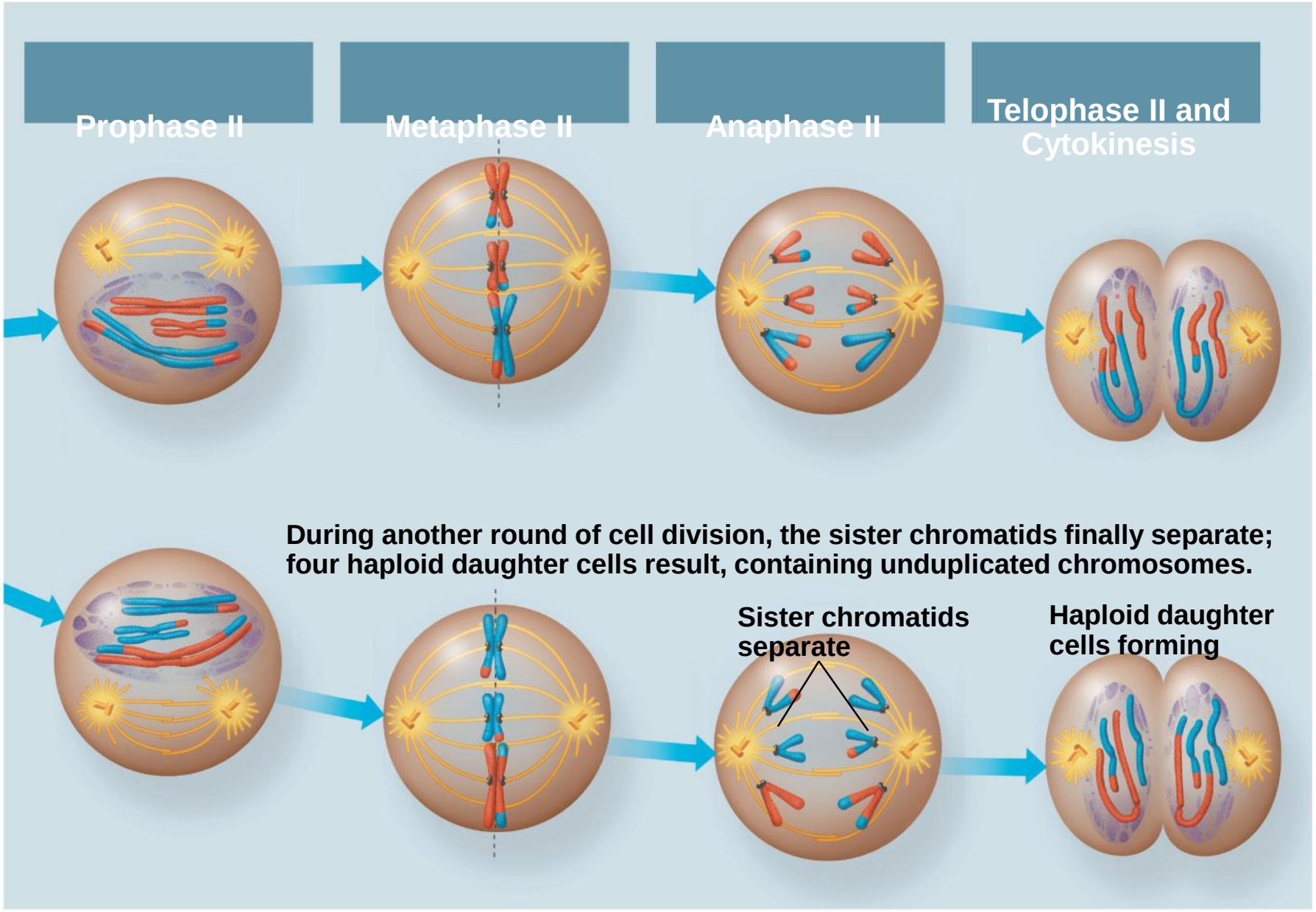
Prophase I

- Prophase I typically occupies more than 90% of the time required for meiosis
- Chromosomes begin to condense
- In **synapsis**, homologous chromosomes loosely pair up, aligned gene by gene

- In **crossing over**, nonsister chromatids exchange DNA segments
- Each pair of chromosomes forms a tetrad, a group of four chromatids
- Each tetrad usually has one or more **chiasmata**, X-shaped regions where crossing over occurred

Metaphase I

- In metaphase I, tetrads line up at the metaphase plate, with one chromosome facing each pole
- Microtubules from one pole are attached to the kinetochore of one chromosome of each tetrad
- Microtubules from the other pole are attached to the kinetochore of the other chromosome



Anaphase I

- In anaphase I, pairs of homologous chromosomes separate
- One chromosome moves toward each pole, guided by the spindle apparatus
- Sister chromatids remain attached at the centromere and move as one unit toward the pole

Telophase I and Cytokinesis

- In the beginning of telophase I, each half of the cell has a haploid set of chromosomes; each chromosome still consists of two sister chromatids
- Cytokinesis usually occurs simultaneously, forming two haploid daughter cells

- In animal cells, a cleavage furrow forms; in plant cells, a cell plate forms
- No chromosome replication occurs between the end of meiosis I and the beginning of meiosis II because the chromosomes are already replicated

- Division in meiosis II also occurs in four phases
  - Prophase II
  - Metaphase II
  - Anaphase II
  - Telophase II and cytokinesis
- Meiosis II is very similar to mitosis

Prophase II

- In prophase II, a spindle apparatus forms
- In late prophase II, chromosomes (each still composed of two chromatids) move toward the metaphase plate

Metaphase II

- In metaphase II, the sister chromatids are arranged at the metaphase plate
- Because of crossing over in meiosis I, the two sister chromatids of each chromosome are no longer genetically identical
- The kinetochores of sister chromatids attach to microtubules extending from opposite poles

Anaphase II

- In anaphase II, the sister chromatids separate
- The sister chromatids of each chromosome now move as two newly individual chromosomes toward opposite poles

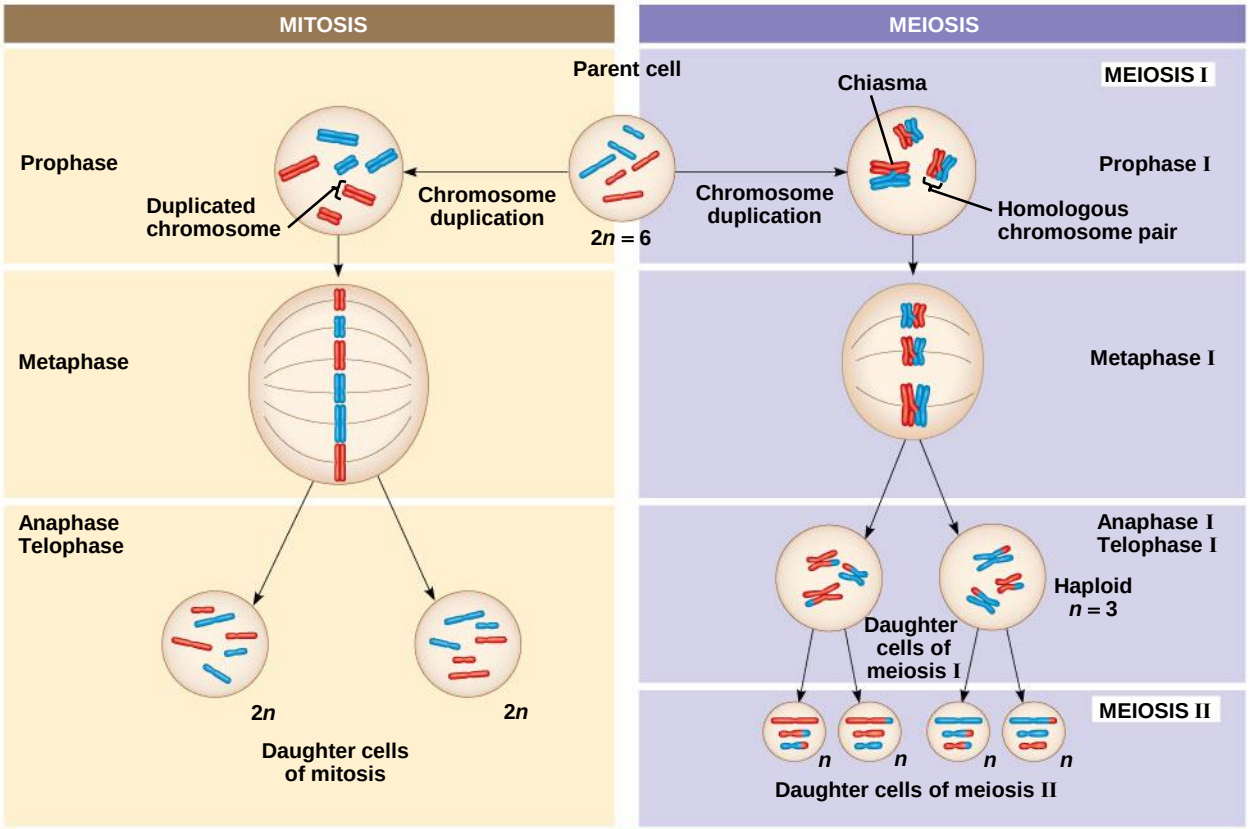
Telophase II and Cytokinesis

- In telophase II, the chromosomes arrive at opposite poles
- Nuclei form, and the chromosomes begin decondensing

- Cytokinesis separates the cytoplasm
- At the end of meiosis, there are four daughter cells, each with a haploid set of unreplicated chromosomes
- Each daughter cell is genetically distinct from the others and from the parent cell

A Comparison of Mitosis and Meiosis

- Mitosis conserves the number of chromosome sets, producing cells that are genetically identical to the parent cell
- Meiosis reduces the number of chromosomes sets from two (diploid) to one (haploid), producing cells that differ genetically from each other and from the parent cell



SUMMARY

Property	Mitosis	Meiosis
DNA replication	Occurs during interphase before mitosis begins	Occurs during interphase before meiosis I begins
Number of divisions	One, including prophase, 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 diploid ($2n$) and genetically identical to the parent cell	Four, each haploid (n), containing half as many chromosomes as the parent cell; genetically different from the parent cell and from each other
Role in the animal body	Enables multicellular adult to arise from zygote; produces cells for growth, repair, and, in some species, asexual reproduction	Produces gametes; reduces number of chromosomes by half and introduces genetic variability among the gametes

Figure 13.9a

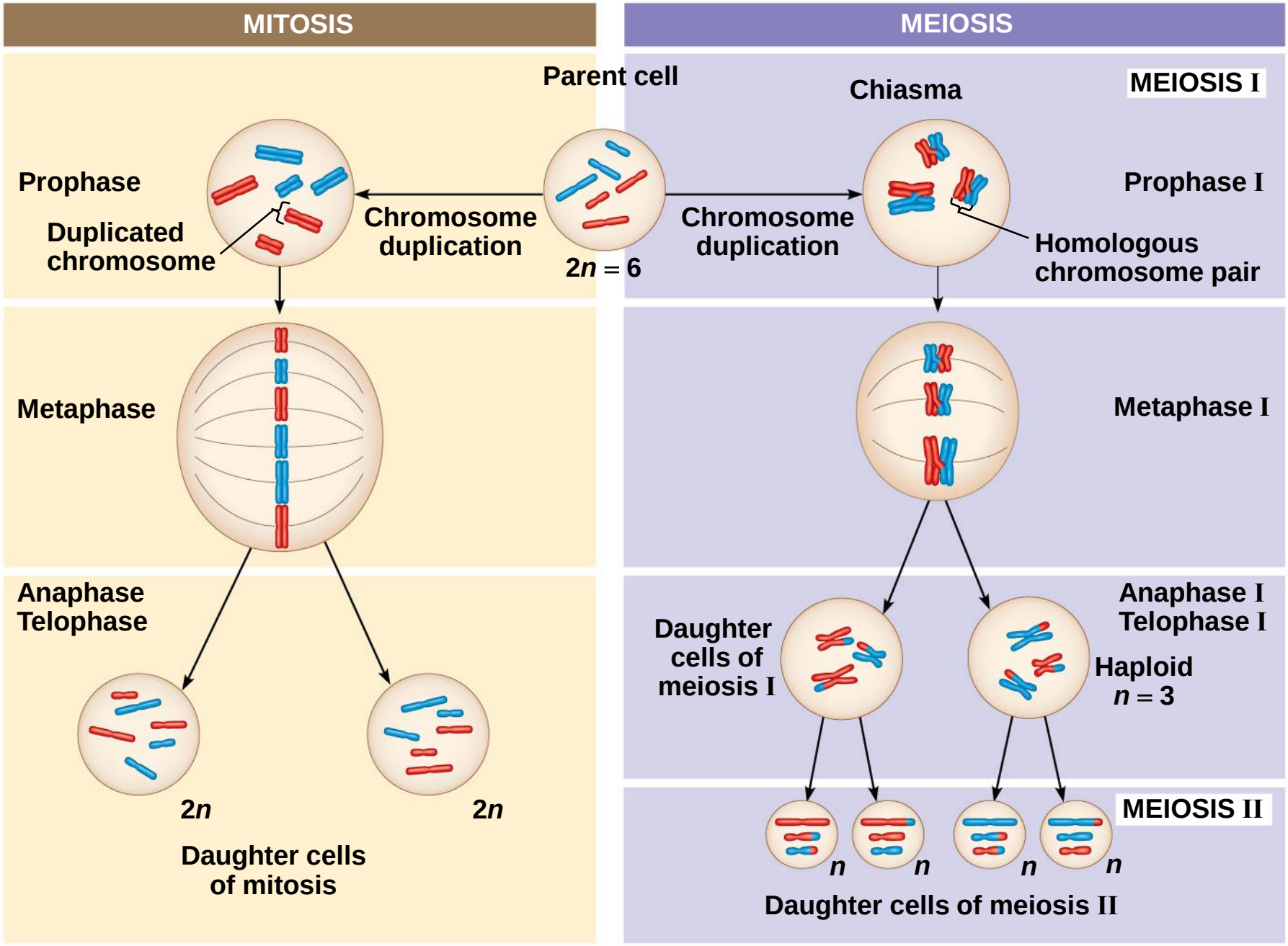


Figure 13.9b

SUMMARY		
Property	**Mitosis**	**Meiosis**
DNA replication	Occurs during interphase before mitosis begins	Occurs during interphase before meiosis I begins
Number of divisions	One, including prophase, metaphase, anaphase, and telophase	Two, each including prophase, metaphase, anaphase, and telophase
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Role in the animal body	Enables multicellular adult to arise from zygote; produces cells for growth, repair, and, in some species, asexual reproduction	Produces gametes; reduces number of chromosomes by half and introduces genetic variability among the gametes

- Three events are unique to meiosis, and all three occur in meiosis l
  - Synapsis and crossing over in prophase I: Homologous chromosomes physically connect and exchange genetic information
  - At the metaphase plate, there are paired homologous chromosomes (tetrads), instead of individual replicated chromosomes
  - At anaphase I, it is homologous chromosomes, instead of sister chromatids, that separate

- Sister chromatid cohesion allows sister chromatids of a single chromosome to stay together through meiosis I
- Protein complexes called cohesins are responsible for this cohesion
- In mitosis, cohesins are cleaved at the end of metaphase
- In meiosis, cohesins are cleaved along the chromosome arms in anaphase I (separation of homologs) and at the centromeres in anaphase II (separation of sister chromatids)

4: Genetic variation produced in sexual life cycles contributes to evolution

- Mutations (changes in an organism's DNA) are the original source of genetic diversity
- Mutations create different versions of genes called alleles
- Reshuffling of alleles during sexual reproduction produces genetic variation

Origins of Genetic Variation Among Offspring: 12th Nov

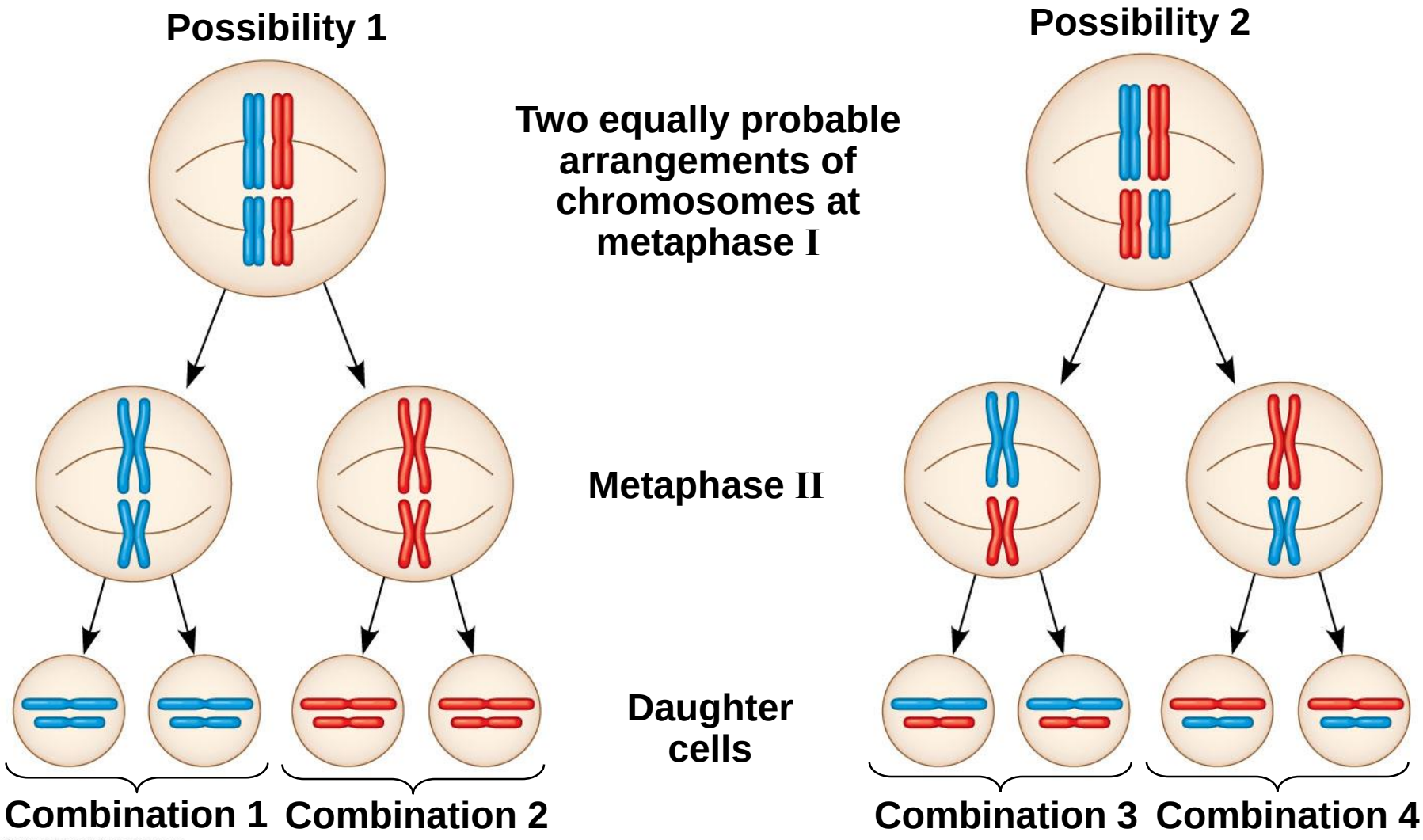
- The behavior of chromosomes during meiosis and fertilization is responsible for most of the variation that arises in each generation
- Three mechanisms contribute to genetic variation
  - Independent assortment of chromosomes
  - Crossing over
  - Random fertilization

Independent Assortment of Chromosomes

- Homologous pairs of chromosomes orient randomly at metaphase I of meiosis
- In independent assortment, each pair of chromosomes sorts maternal and paternal homologues into daughter cells independently of the other pairs

- The number of combinations possible when chromosomes assort independently into gametes is 2^n, where *n* is the haploid number
- For humans (*n* = 23), there are more than 8 million (2^{23}) possible combinations of chromosomes

Figure 13.10-3



Crossing Over

- Crossing over produces **recombinant chromosomes**, which combine DNA inherited from each parent
- Crossing over begins very early in prophase I, as homologous chromosomes pair up gene by gene

- In crossing over, homologous portions of two nonsister chromatids trade places
- Crossing over contributes to genetic variation by combining DNA from two parents into a single chromosome

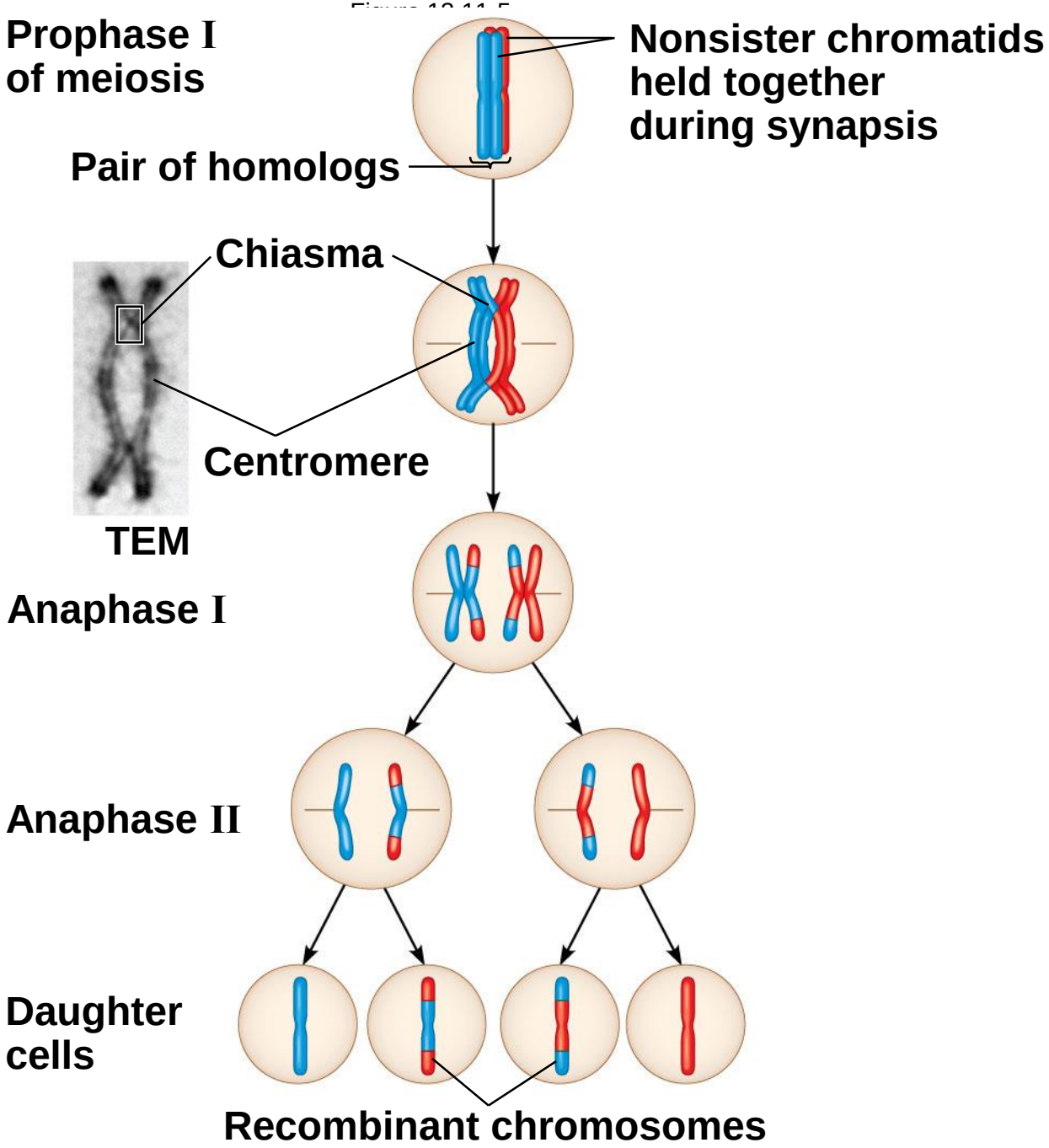
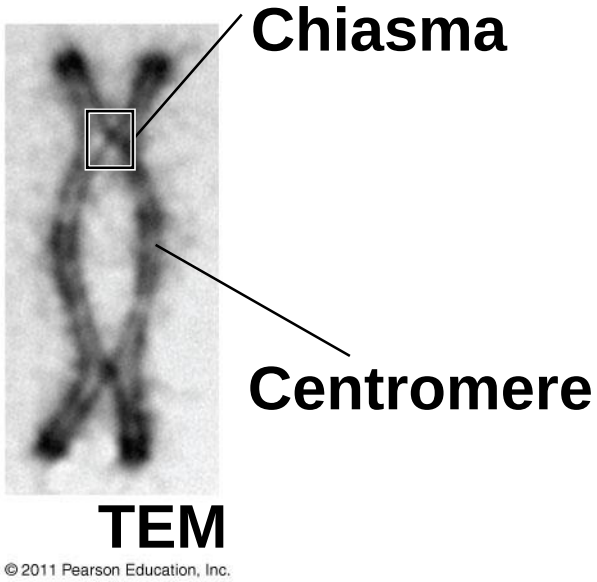


Figure 13.11a



Random Fertilization

- Random fertilization adds to genetic variation because any sperm can fuse with any ovum (unfertilized egg)
- The fusion of two gametes (each with 8.4 million possible chromosome combinations from independent assortment) produces a zygote with any of about 70 trillion diploid combinations

- Crossing over adds even more variation
- Each zygote has a unique genetic identity

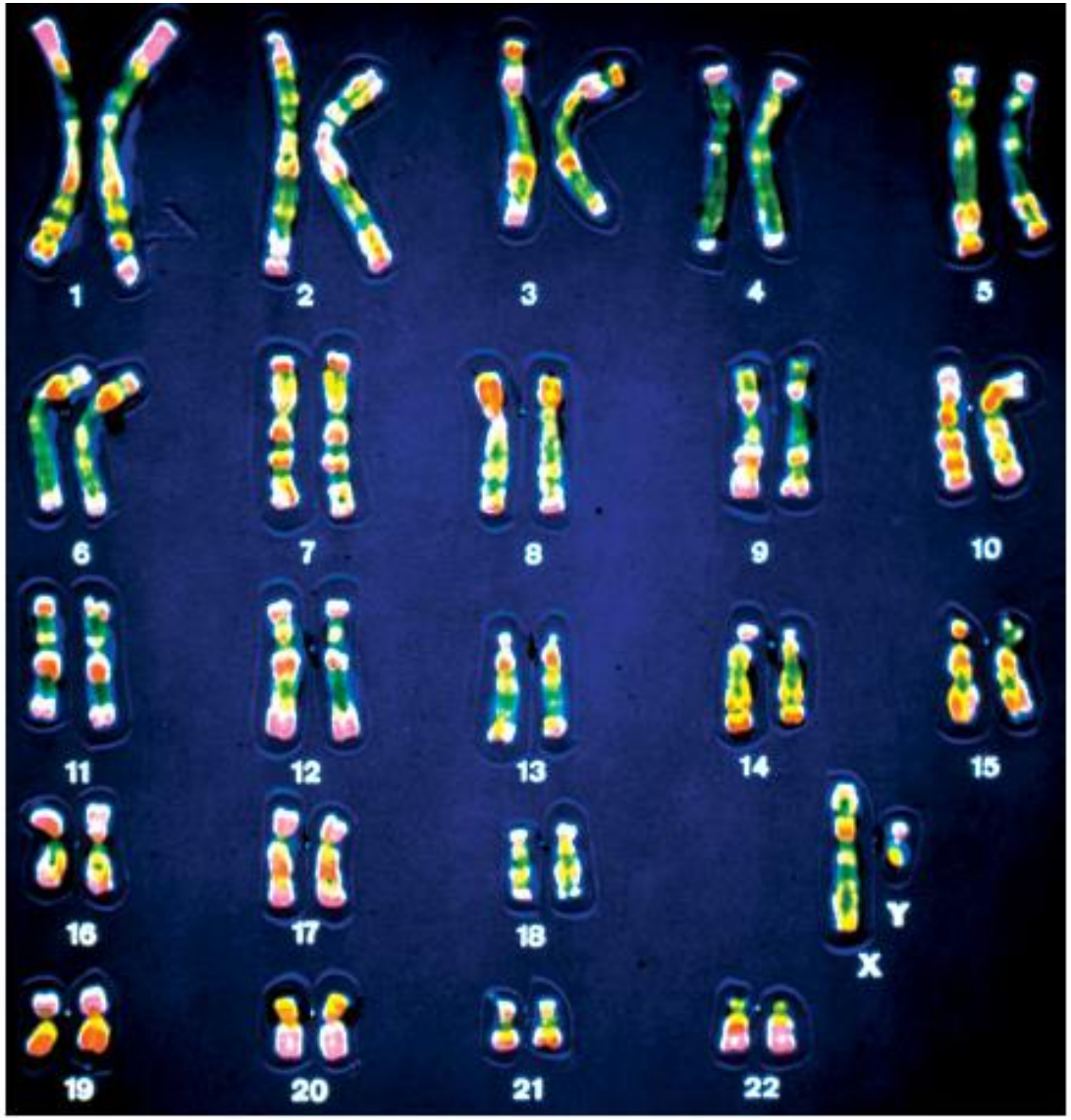
The Evolutionary Significance of Genetic Variation Within Populations

- Natural selection results in the accumulation of genetic variations favored by the environment
- Sexual reproduction contributes to the genetic variation in a population, which originates from mutations

Chromosomes and Heredity

- Heredity = transmission of genetic characteristics from parent to offspring
- Karyotype = chart of chromosomes at metaphase
- Humans have 23 pairs homologous chromosomes in somatic cells (diploid number)
  - 1 chromosome inherited from each parent
  - 22 pairs called autosomes
  - one pair of sex chromosomes (X and Y)
    - normal female has 2 X chromosomes
    - normal male has one X and one Y chromosome
- Sperm and egg cells contain 23 haploid chromosomes
  - paternal chromosomes combine with maternal

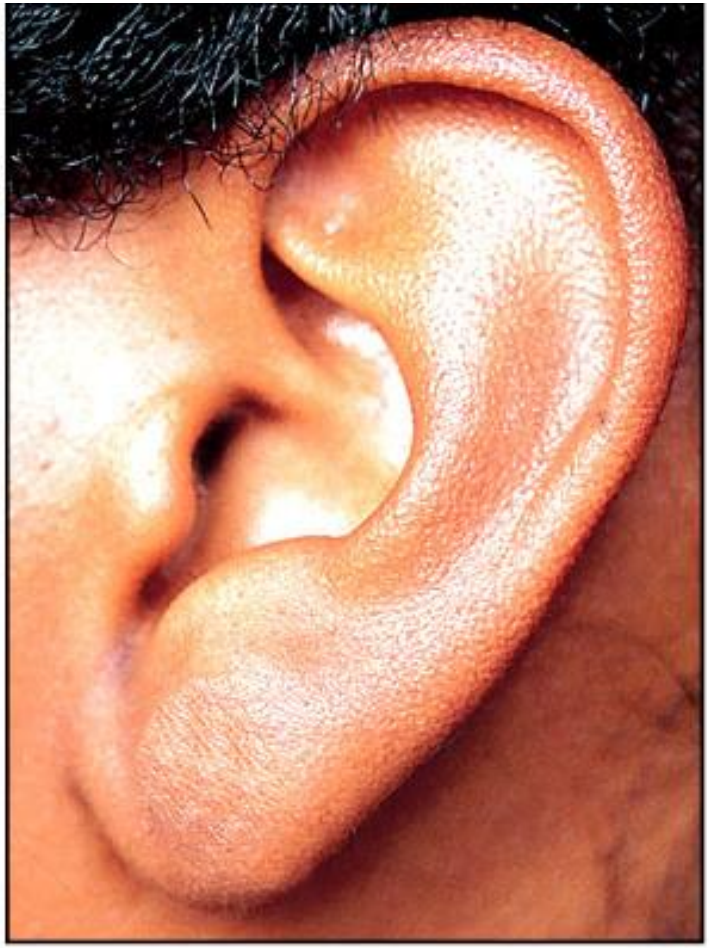
Karyotype of Normal Human Male



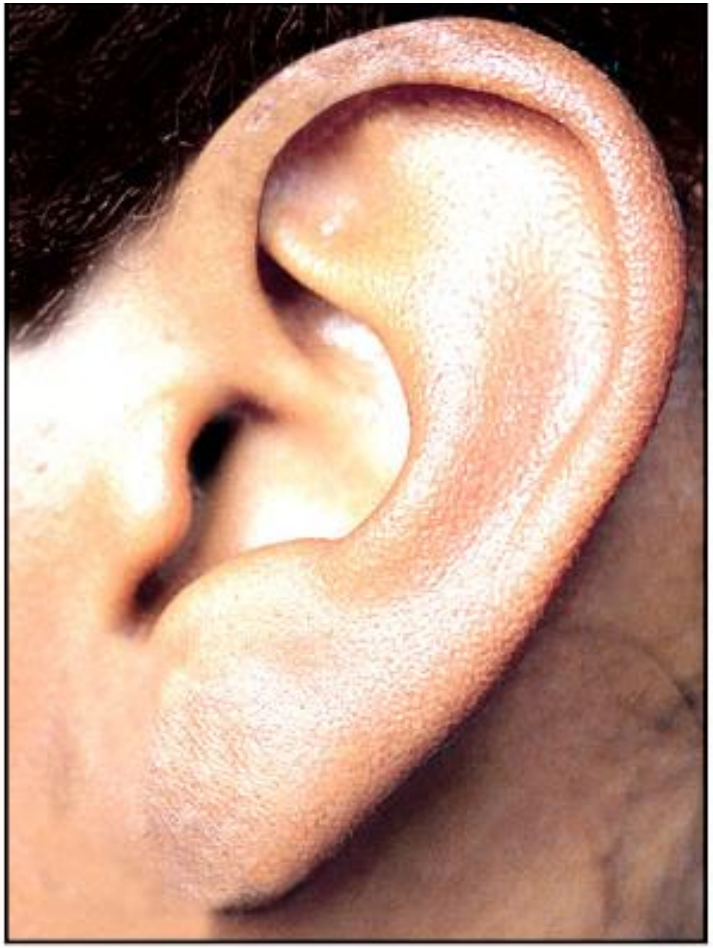
Genes and Alleles

- Gene loci
  - location of gene on chromosome
- Alleles
  - different forms of gene at same locus on 2 homologous chromosomes
- Dominant allele
  - produces protein responsible for visible trait
- Recessive allele
  - expressed only when both alleles are recessive
  - ususually produces abnormal protein variant

Genetics of Earlobes



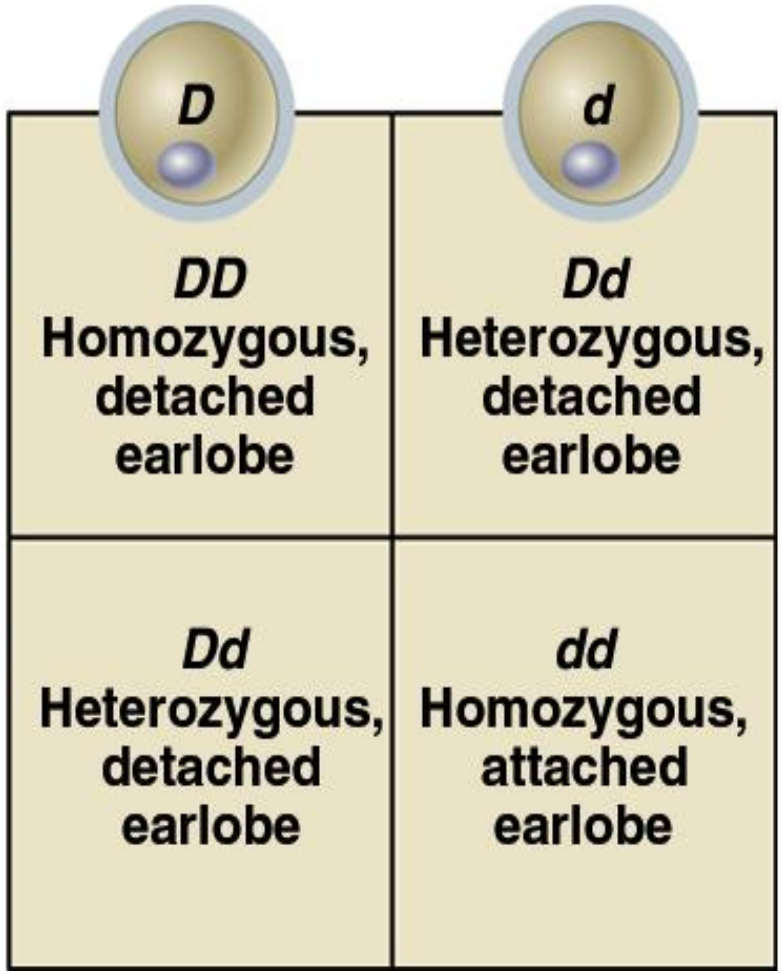
Detached earlobe
DD, Dd



Attached earlobe
dd

Genetics of Earlobes

- Genotype
  - alleles for a particular trait (DD)
- Phenotype
  - trait that results (appearance)
- Dominant allele (D)
  - expressed with DD or Dd
  - Dd parent ‘carrier’ of recessive gene
- Recessive allele (d)
  - expressed with dd only
- Heterozygous carriers of hereditary disease
  - cystic fibrosis

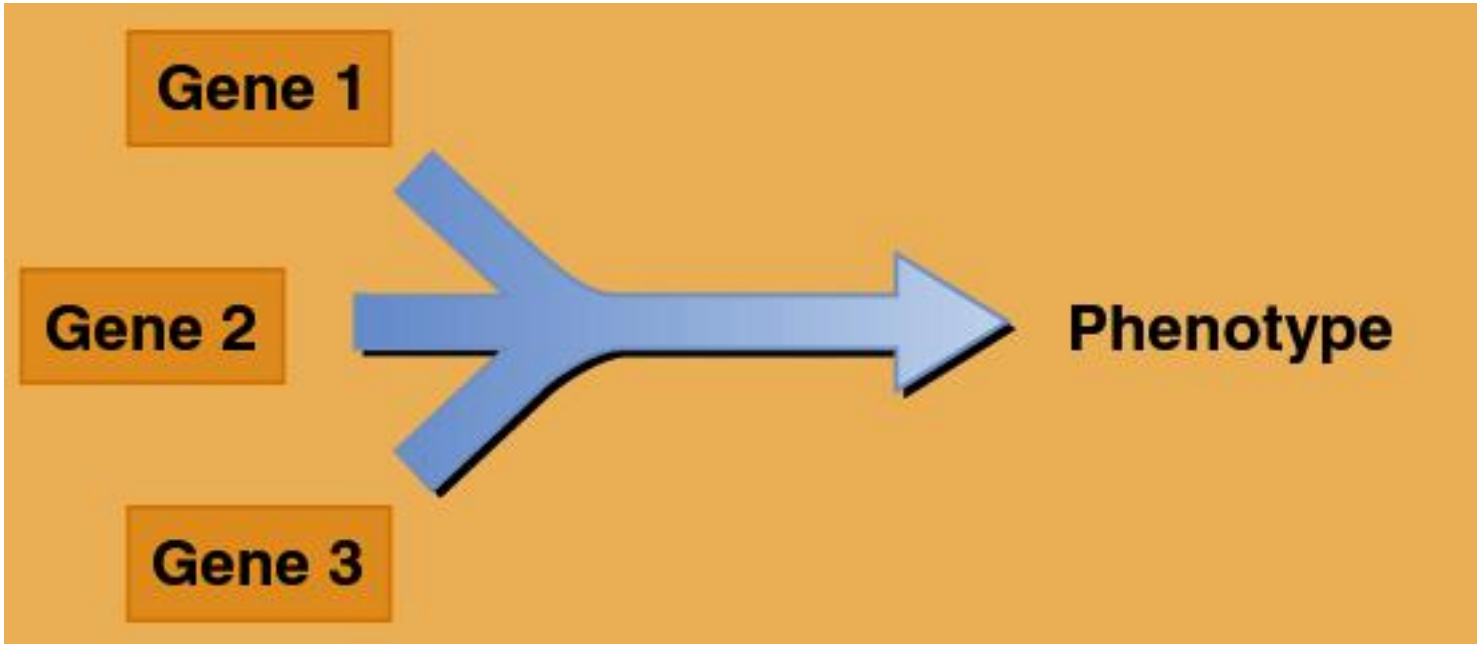


Punnett square

Multiple Alleles, Codominance, Incomplete Dominance

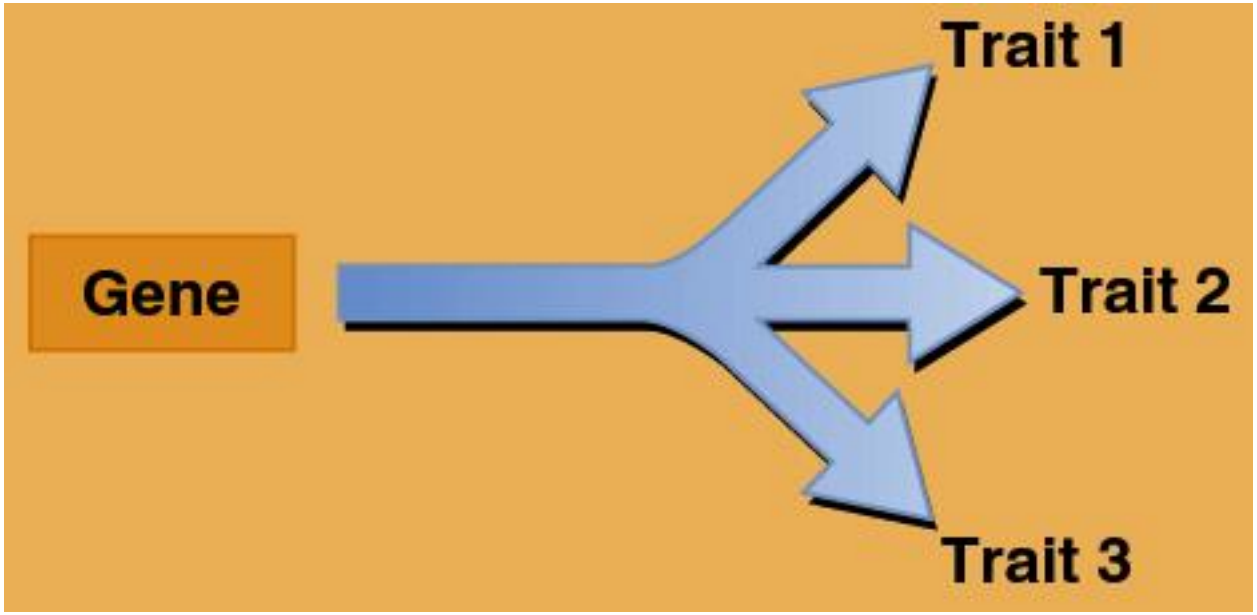
- Gene pool
  - collective genetic makeup of whole population
- Multiple alleles
  - more than 2 alleles for a trait
  - such as I^A, I^B, i alleles for blood type
- Codominant
  - both alleles expressed, I^AI^B = type AB blood
- Incomplete dominance
  - phenotype intermediate between traits for each allele

Polygenic Inheritance



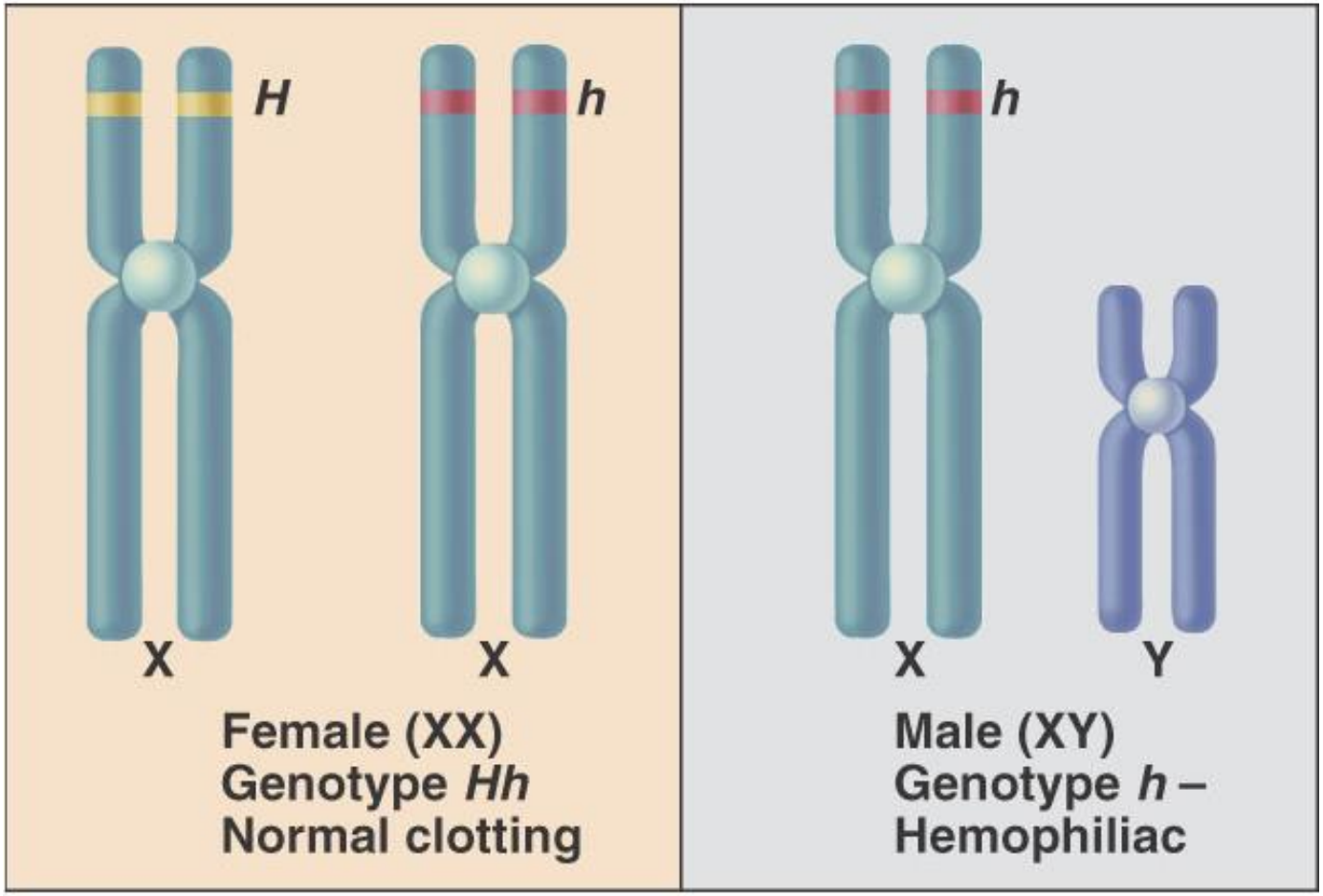
- 2 or more genes combine their effects to produce single phenotypic trait, such as skin and eye color, alcoholism and heart disease

Pleiotropy



- Single gene causes multiple phenotypic traits (ex. sickle-cell disease)
  - sticky, fragile, abnormal shaped red blood cells at low oxygen levels cause anemia and enlarged spleen

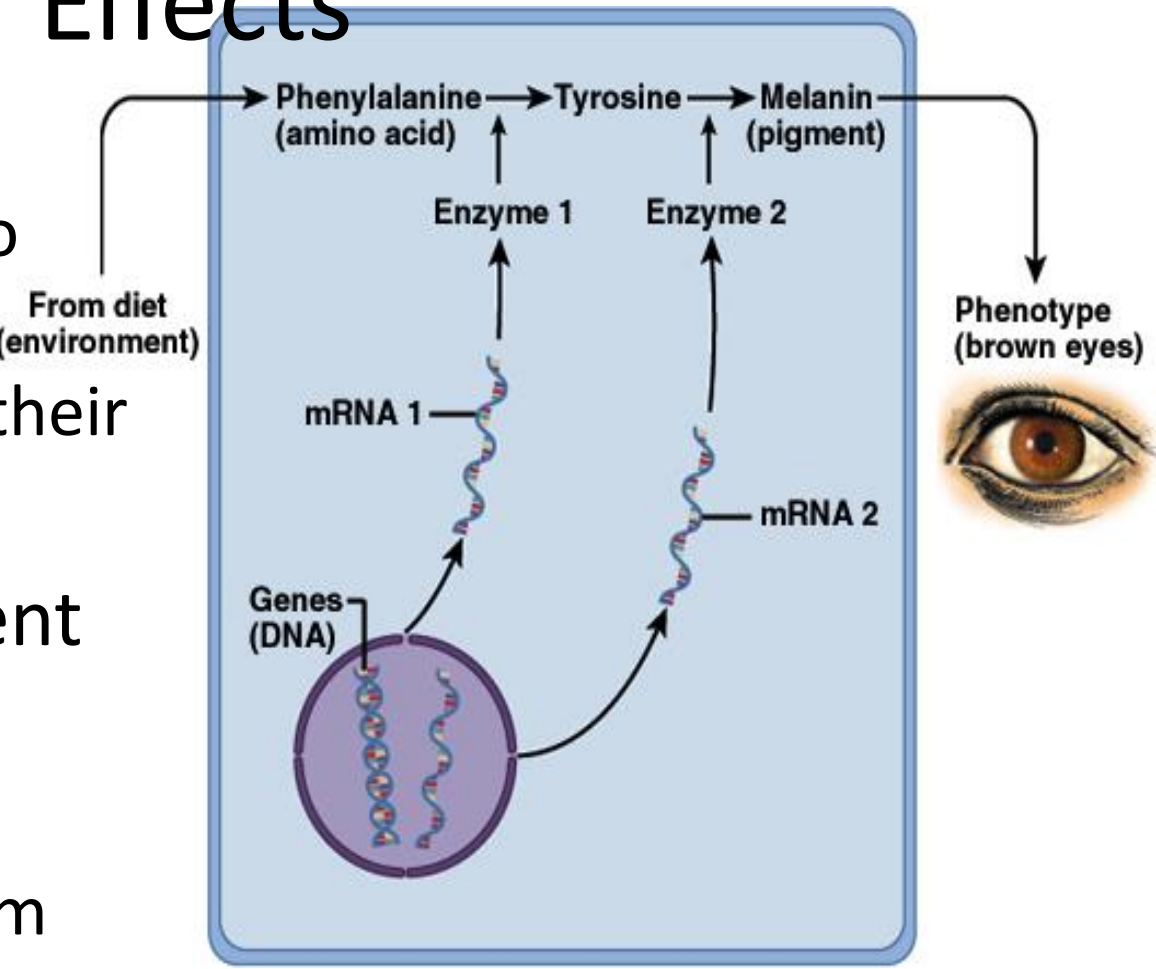
Sex-Linked Inheritance



- Recessive allele on X, no gene locus for trait on Y, so hemophilia more common in men (mother must be carrier)

Penetrance and Environmental Effects

- Penetrance
  - % of population to express predicted phenotype given their genotypes
- Role of environment
  - brown eye color requires phenylalanine from diet to produce melanin, the eye pigment



Alleles at the Population Level

- Dominance and recessiveness of allele do not determine frequency in a population
- Some recessive alleles, blood type O, are the most common
- Some dominant alleles, polydactyly and blood type AB, are rare

Cancer

- Tumors (neoplasms)
  - abnormal growth, when cells multiply faster than they die
  - oncology is the study of tumors
- Benign
  - connective tissue capsule, grow slowly, stays local
  - potentially lethal by compression of vital tissues
- Malignant
  - unencapsulated, fast growing, metastatic (causes 90% of cancer deaths)

Causes of Cancer

- Carcinogens - estimates of 60 - 70% of cancers from environmental agents
  - chemical
    - cigarette tar, food preservatives
  - radiation
    - UV radiation, α particles, γ rays, β particles
  - viruses
    - type 2 herpes simplex - uterus, hepatitis C - liver

Mutagens

- Trigger gene mutations
  - cell may die, be destroyed by immune system or produce a tumor

<u>Defenses against mutagens</u>

- Scavenger cells
  - remove them before they cause genetic damage
- Peroxisomes
  - neutralize nitrites, free radicals and oxidizing agents
- Nuclear enzymes
  - repair DNA
- Tumor necrosis factor (TNF) from macrophages and certain WBCs destroys tumors

Malignant Tumor (Cancer) Genes

- Oncogenes
  - mutated form of normal growth factor genes called proto-oncogenes
  - sis oncogene causes excessive production of growth factors
    - stimulate neovascularization of tumor
  - ras oncogene codes for abnormal growth factor receptors
    - sends constant divide signal to cell
- Tumor suppressor genes
  - inhibit development of cancer
  - damage to one or both removes control of cell division

Effects of Malignancies

- Displaces normal tissue, organ function deteriorates
  - rapid cell growth of immature nonfunctional cells
  - metastatic cells have different tissue origin
- Block vital passageways
  - block air flow and compress or rupture blood vessels
- Diverts nutrients from healthy tissues
  - tumors have high metabolic rates
  - causes weakness, fatigue, emaciation, susceptibility to infection
  - cachexia is extreme wasting away of muscle and adipose tissue