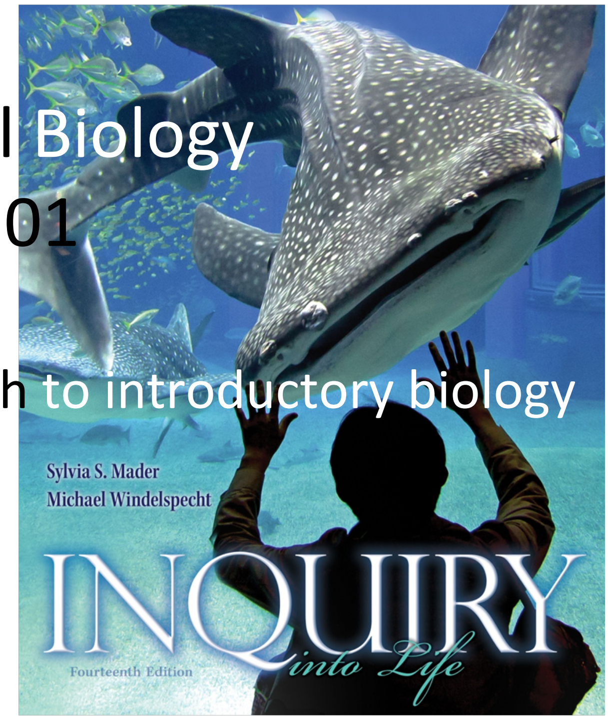
Fundamental Biology
BI 1101



an interdisciplinary approach to introductory biology

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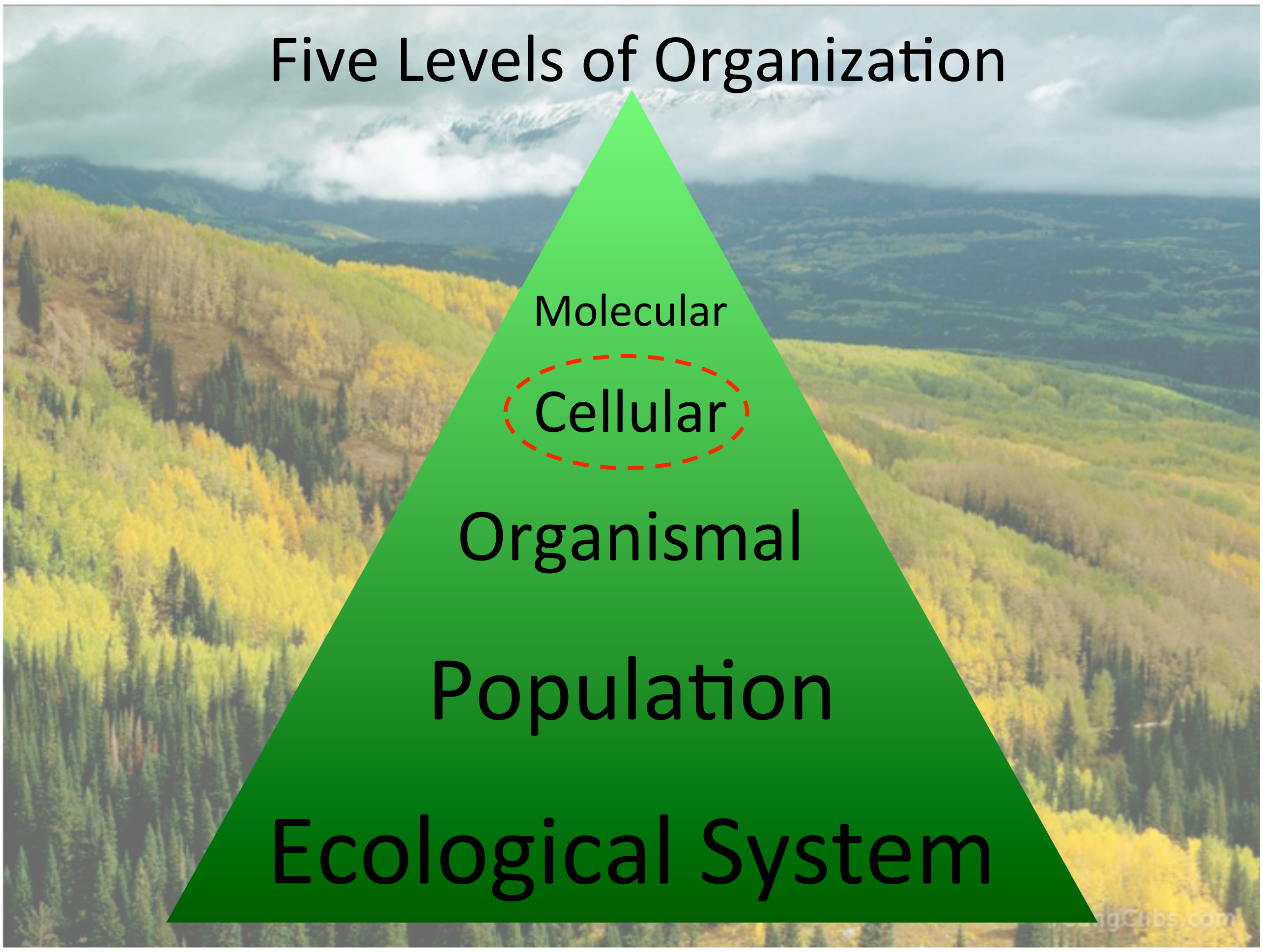
Five Levels of Organization



CELLULAR BASIS OF REPRODUCTION AND INHERITANCE

Learning outcome

After this chapter, students are able to:

- Describe the cellular basic of reproduction and inheritance
- Explain basic structure of chromososme
- Compare cell cycle, mitosis and meiosis

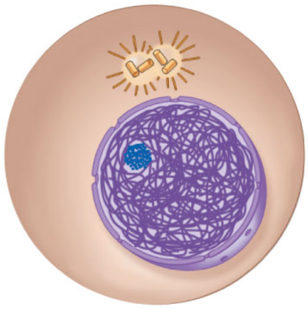
- Rain Forest Rescue
  - Scientists in Hawaii
    - Have attempted to “rescue” endangered species from extinction
  - The goals of these scientists were
    - To promote reproduction to produce more individuals of specific endangered plants

Cyanea kuhihewa

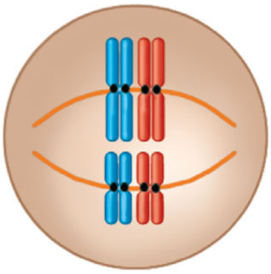
  - In sexual reproduction
    - Fertilization of sperm and egg produces offspring
  - In asexual reproduction
    - Offspring are produced by a single parent, without the participation of sperm and egg

Big Ideas

Cell Division and Reproduction



The Eukaryotic Cell Cycle and Mitosis



Meiosis and Crossing Over

Alterations of Chromosome Number and Structure

CELL DIVISION AND REPRODUCTION

Cell division plays many important roles in the lives of organisms

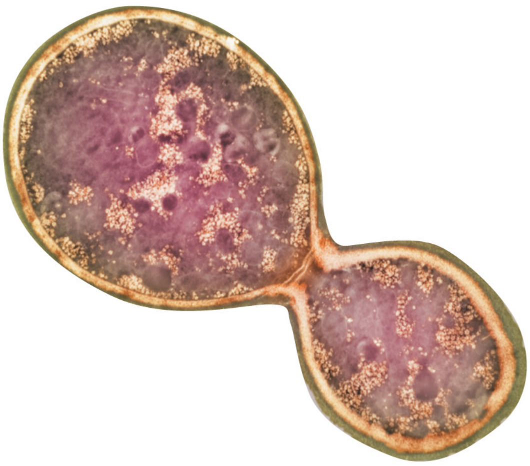
- Organisms reproduce their own kind, a key characteristic of life.
- Cell division
  - is reproduction at the cellular level,
  - requires the duplication of chromosomes, and
  - sorts new sets of chromosomes into the resulting pair of daughter cells.

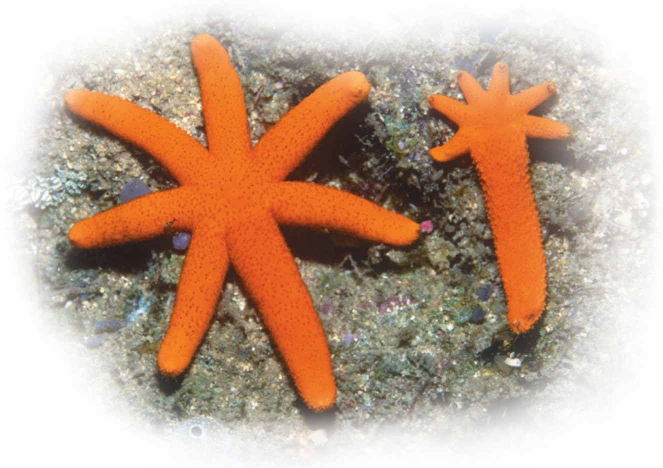
Cell division plays many important roles in the lives of organisms

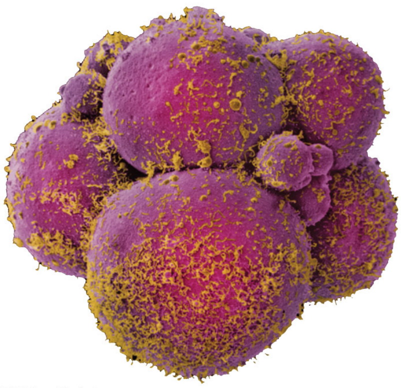
- Cell division is used
  - for reproduction of single-celled organisms,
  - growth of multicellular organisms from a fertilized egg into an adult,
  - repair and replacement of cells, and
  - sperm and egg production.

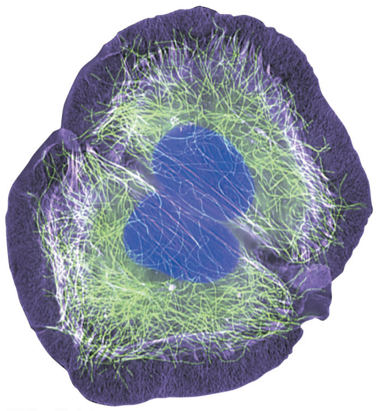
Cell division plays many important roles in the lives of organisms

- Living organisms reproduce by two methods.
  - **Asexual reproduction**
    - produces offspring that are identical to the original cell or organism and
    - involves inheritance of all genes from one parent.
  - **Sexual reproduction**
    - produces offspring that are similar to the parents, but show variations in traits and
    - involves inheritance of unique sets of genes from two parents.









The large, complex chromosomes of eukaryotes duplicate with each cell division

- Eukaryotic cells
  - are more complex and larger than prokaryotic cells,
  - have more genes, and
  - store most of their genes on multiple chromosomes within the nucleus.

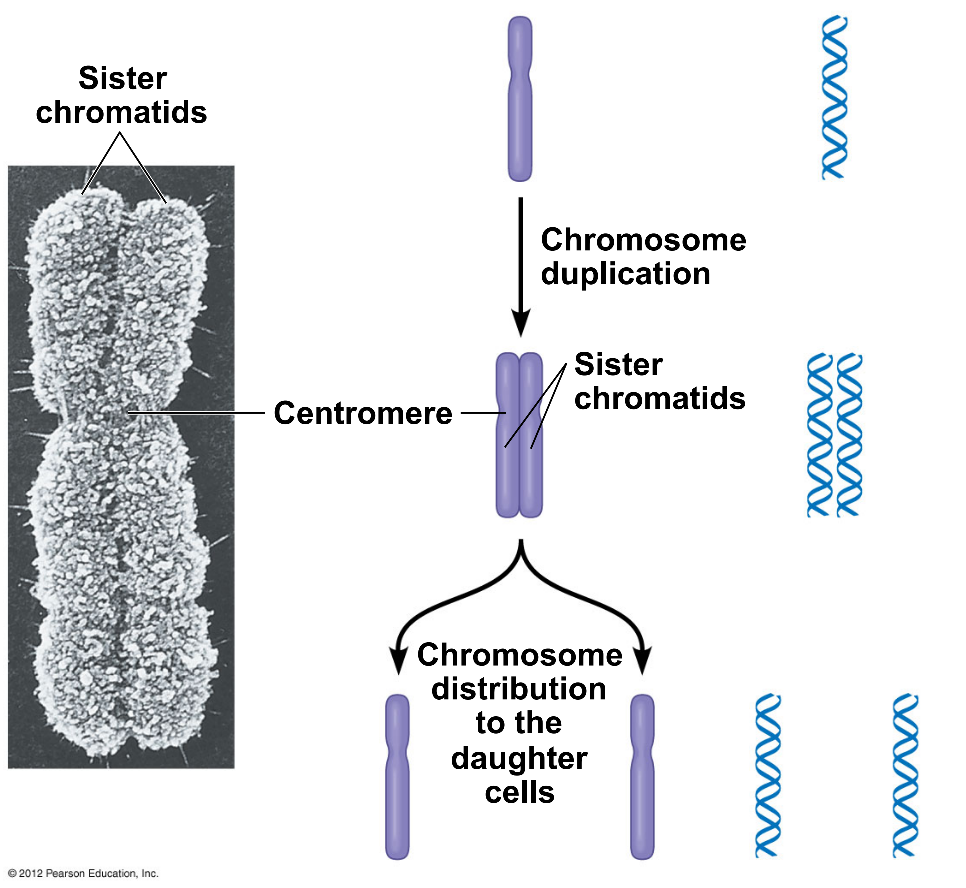
The large, complex chromosomes of eukaryotes duplicate with each cell division

- Eukaryotic chromosomes are composed of **chromatin** consisting of
  - one long DNA molecule and
  - proteins that help maintain the chromosome structure and control the activity of its genes.
- To prepare for division, the chromatin becomes
  - highly compact and
  - visible with a microscope.

The large, complex chromosomes of eukaryotes duplicate with each cell division

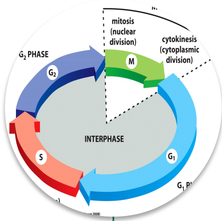
- Before a eukaryotic cell begins to divide, it duplicates all of its chromosomes, resulting in
  - two copies called **sister chromatids**
  - joined together by a narrowed "waist" called the **centromere**.
- When a cell divides, the sister chromatids
  - separate from each other, now called chromosomes, and
  - sort into separate daughter cells.

Figure 8.3B



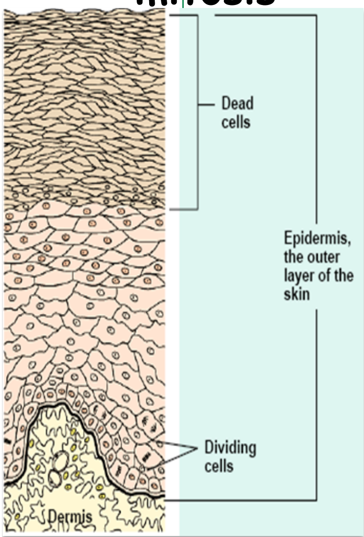
Eukaryotic cells divide in one of two ways

Eukaryotic cell reproduction



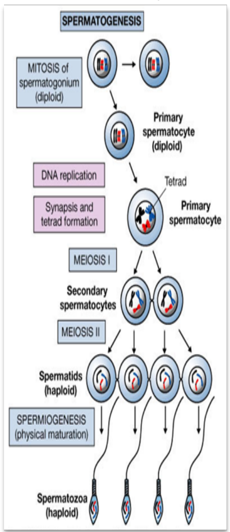
In eukaryotes, nuclei divide by either: mitosis or meiosis.

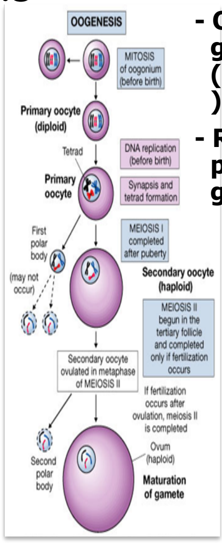
mitosis



- Occurs in somatic (nonreproductive) cells
- Asexual reproduction, development, growth and cell replacement are mitotic divisions

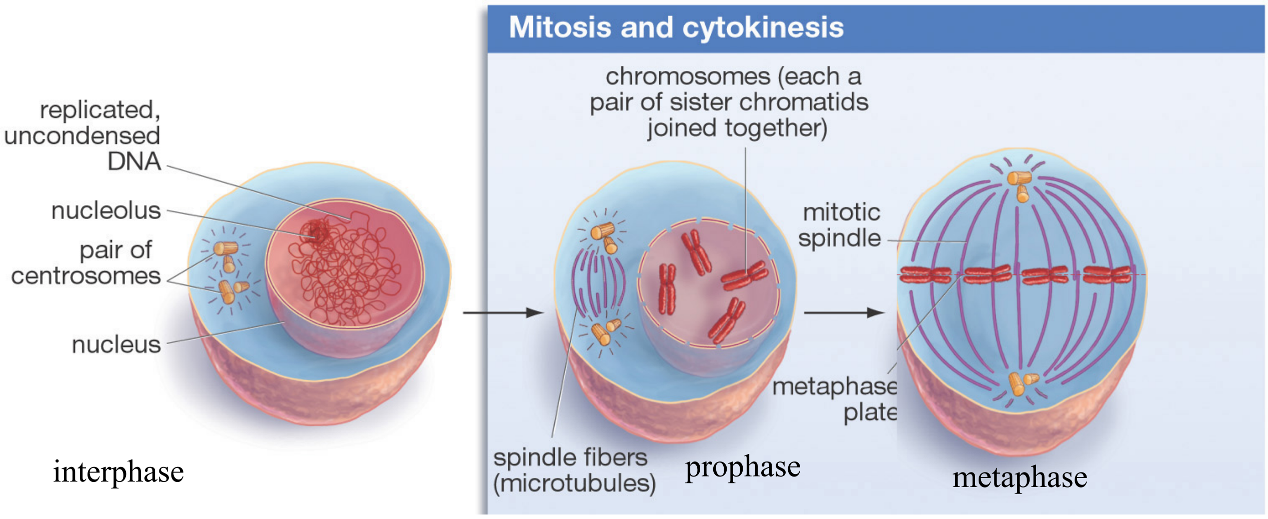
meiosis





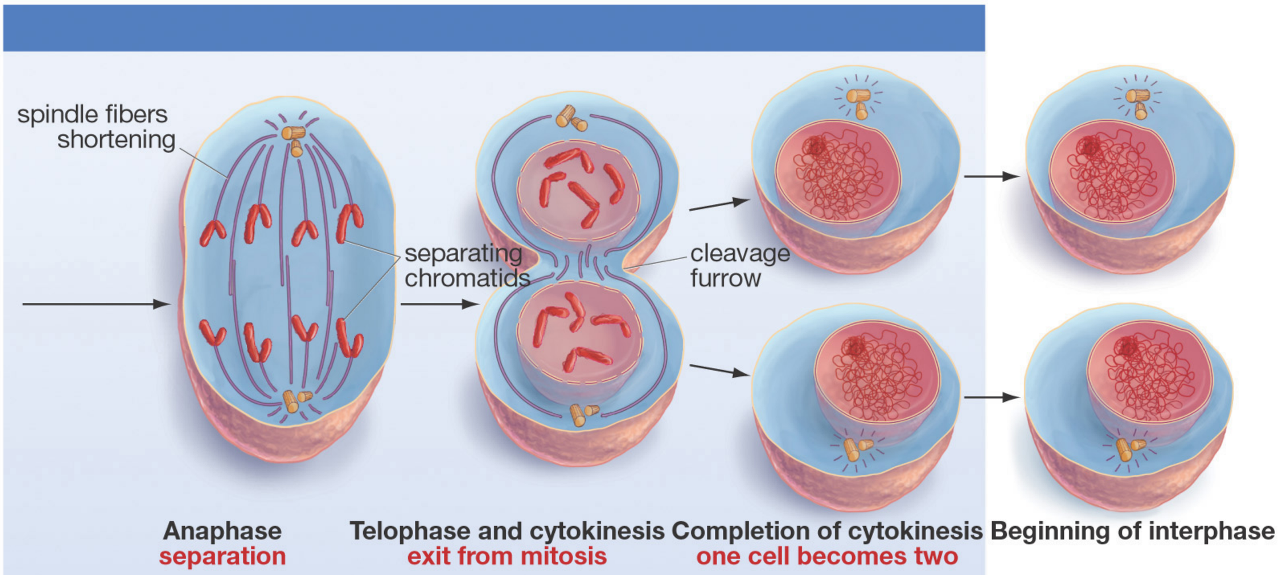
- Occurs in germ (reproductive) cells
- Results in the production of gametes

The Knit of Identity - Mitosis Precisely and Evenly Divides Duplicated Chromosomes

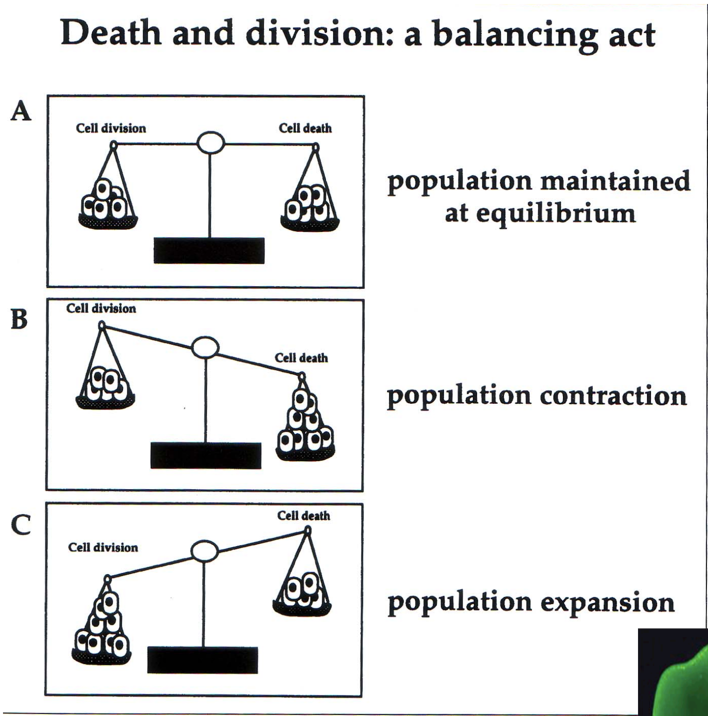


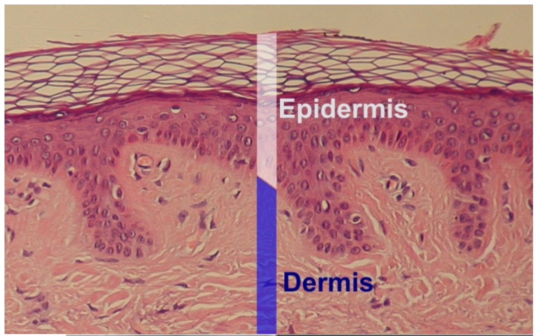
Precisely dividing the duplicated chromosomes has the consequence of providing each new cell with an identical and complete set of genetic instructions.

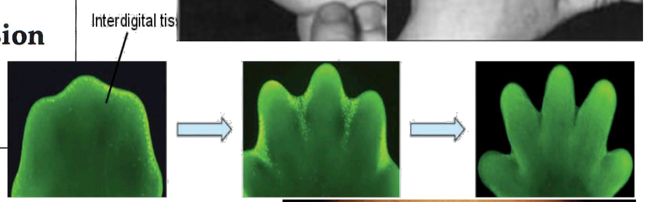
Mitosis Precisely and Evenly Divides Duplicated Chromosomes



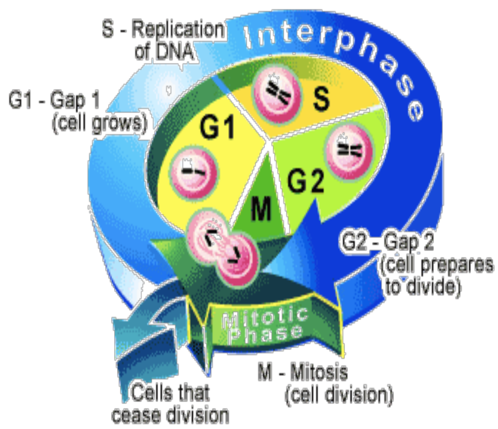
Cytokinesis is the process of cell division and it is distinct and separable from mitosis. Cell division is necessary for reproduction, growth, and repair of an organism.



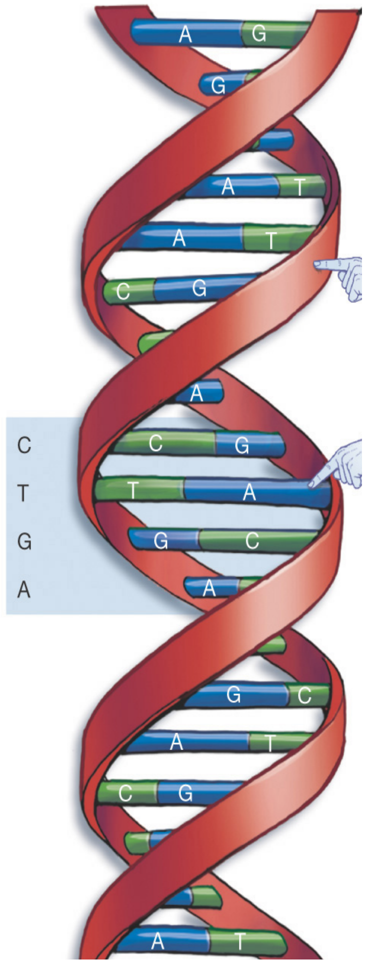




Programmed Cell death – apoptosis



Cell cycle



Cell cycle

- The most basic function of the cell cycle is to **duplicate** accurately the vast amount of DNA in the chromosomes and then **segregate** the copies precisely into two genetically identical daughter cells.
- The eucaryotic cell cycle is divided into four sequential phases: G_1, **S**, G_2, and **M**.
- During most of the cell cycle, the cell is in interphase, which is divided into three subphases : S, G1, and G2.
- **two major** *phases* of the cell cycle: S and M
  1) **DNA duplication occurs during *S phase*** (S for synthesis)→ 10–12 hours (about half of the cell-cycle time in a typical mammalian cell).
  2) After S phase, **chromosome segregation and cell division occur in *M phase*** (M for *m*itosis)→less time (less than an hour in a mammalian cell). M phase involves a series of dramatic events that begin with nuclear division, or mitosis.

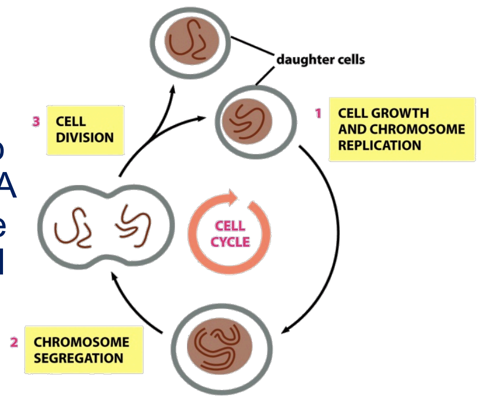


Figure 17-1 Molecular Biology of the Cell 5/e (© Garland Science 2008)

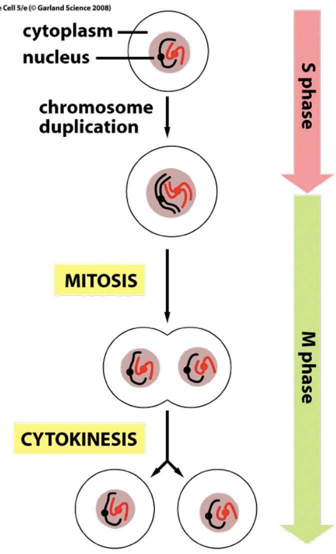
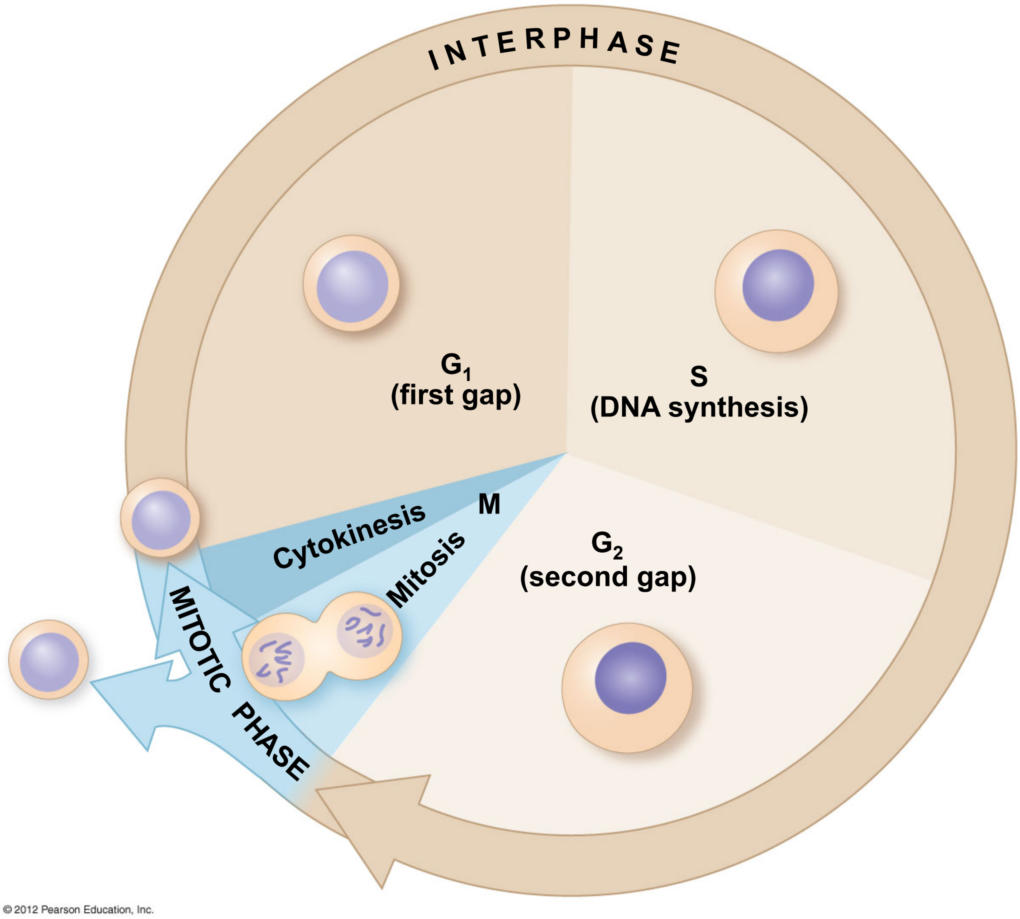


Figure 17-2 Molecular Biology of the Cell 5/e (© Garland Science 2008)

Figure 8.4



The cell cycle multiplies cells

- The cell cycle consists of two stages, characterized as follows:
  1. **Interphase**: duplication of cell contents
     – G_1—growth, increase in cytoplasm
     – S—duplication of chromosomes
     – G_2—growth, preparation for division
  2. **Mitotic phase**: division
     – **Mitosis**—division of the nucleus
     – **Cytokinesis**—division of cytoplasm

Cell division is a continuum of dynamic changes

- Mitosis progresses through a series of stages:
  - prophase,
  - prometaphase,
  - metaphase,
  - anaphase, and
  - telophase.
- Cytokinesis often overlaps telophase.

Figure 8.5_1

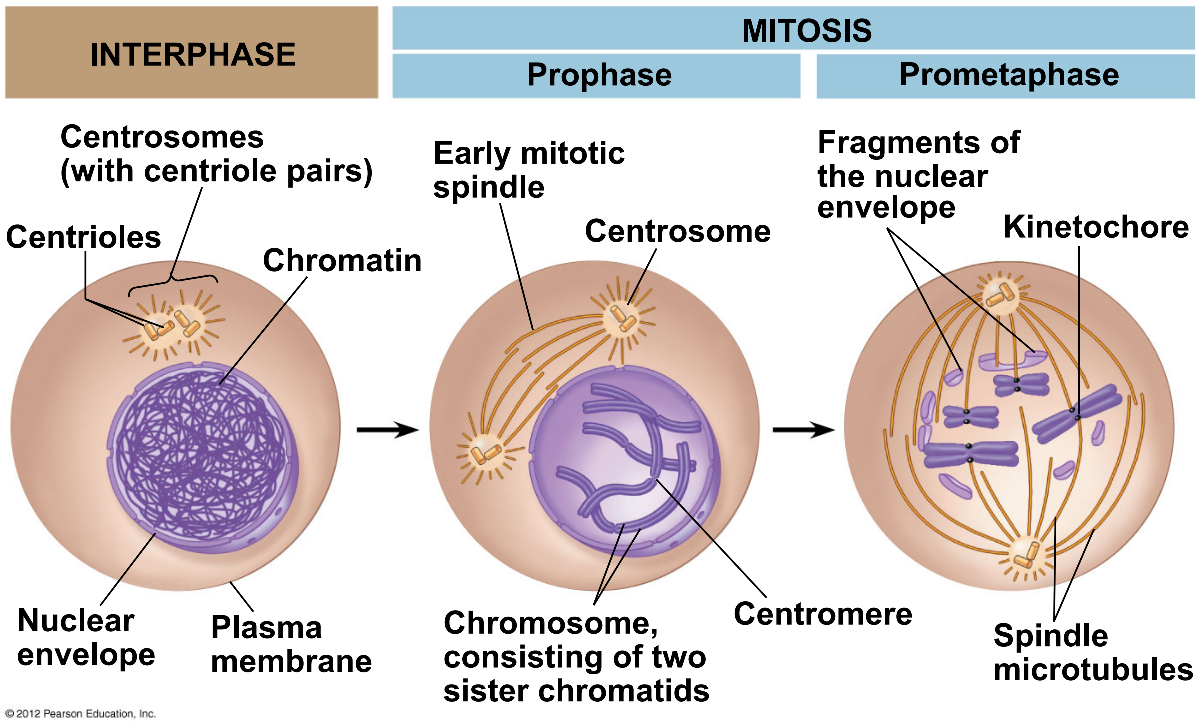


Figure 8.5_left

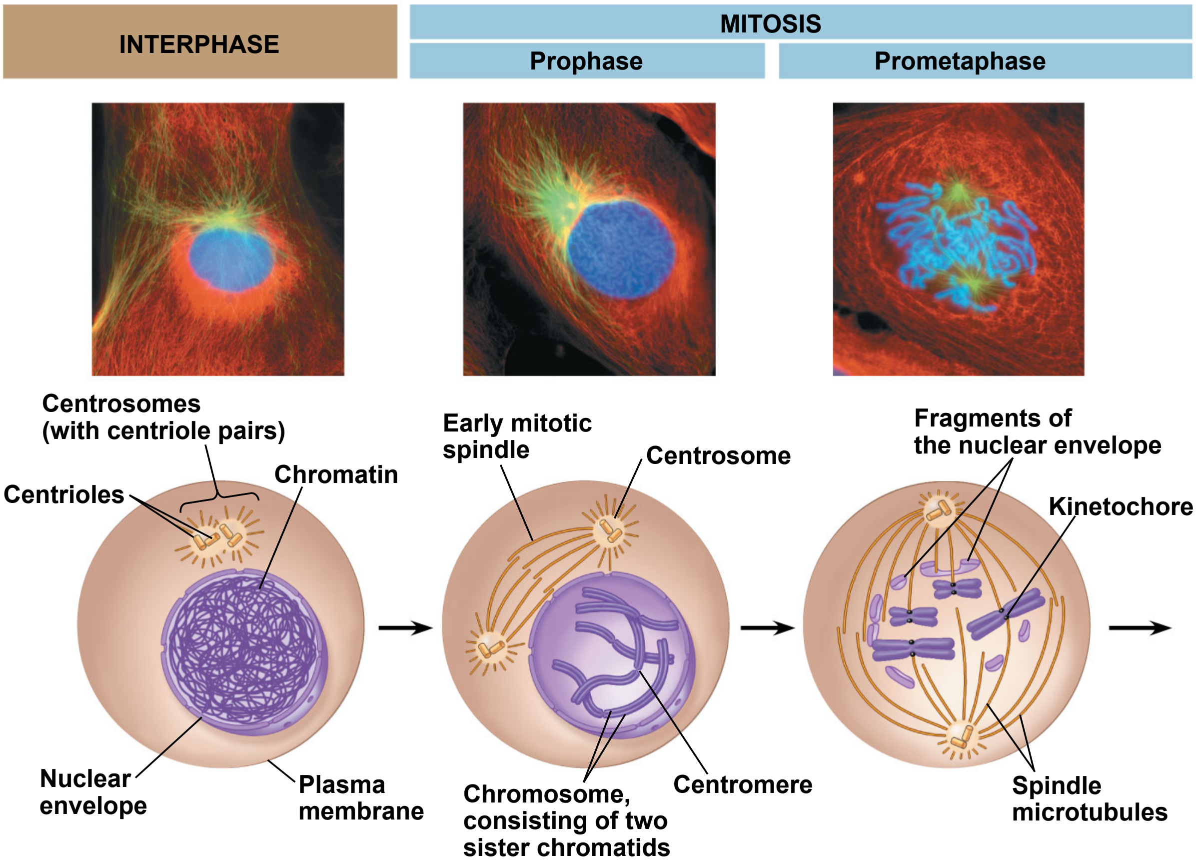
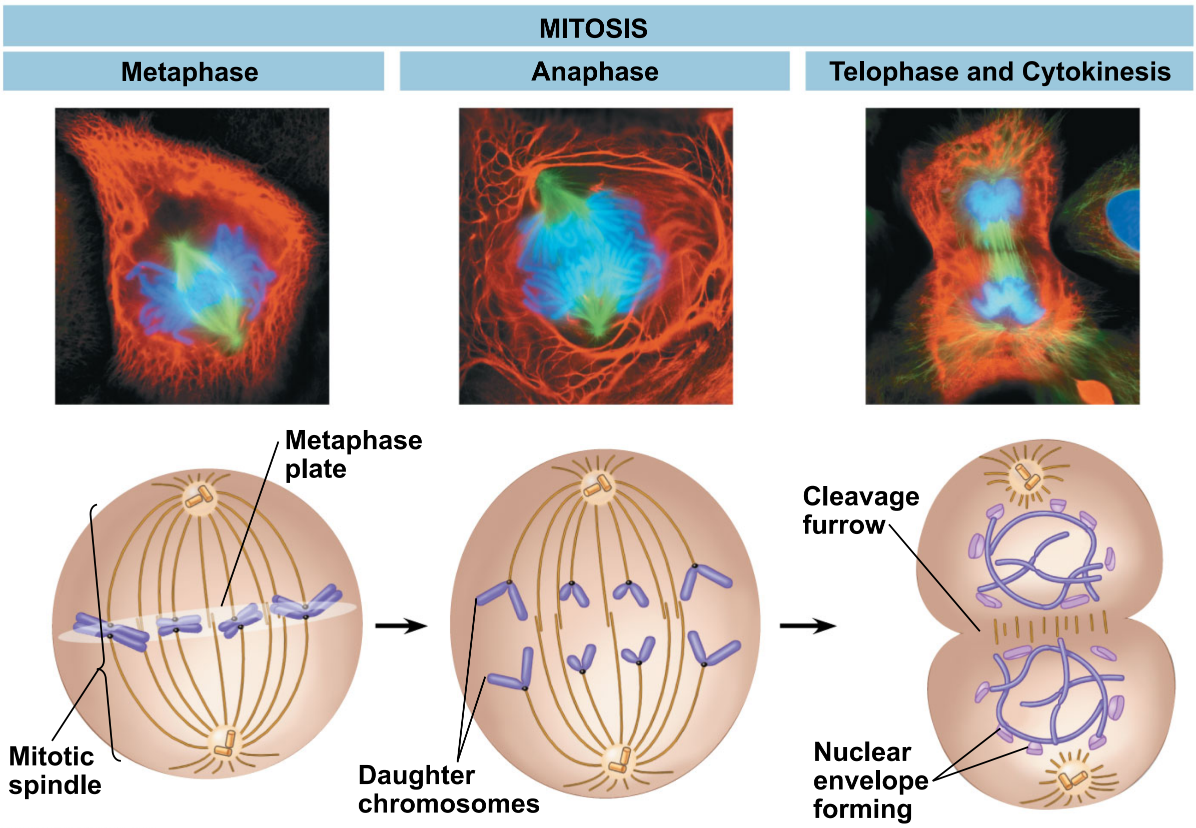


Figure 8.5_right



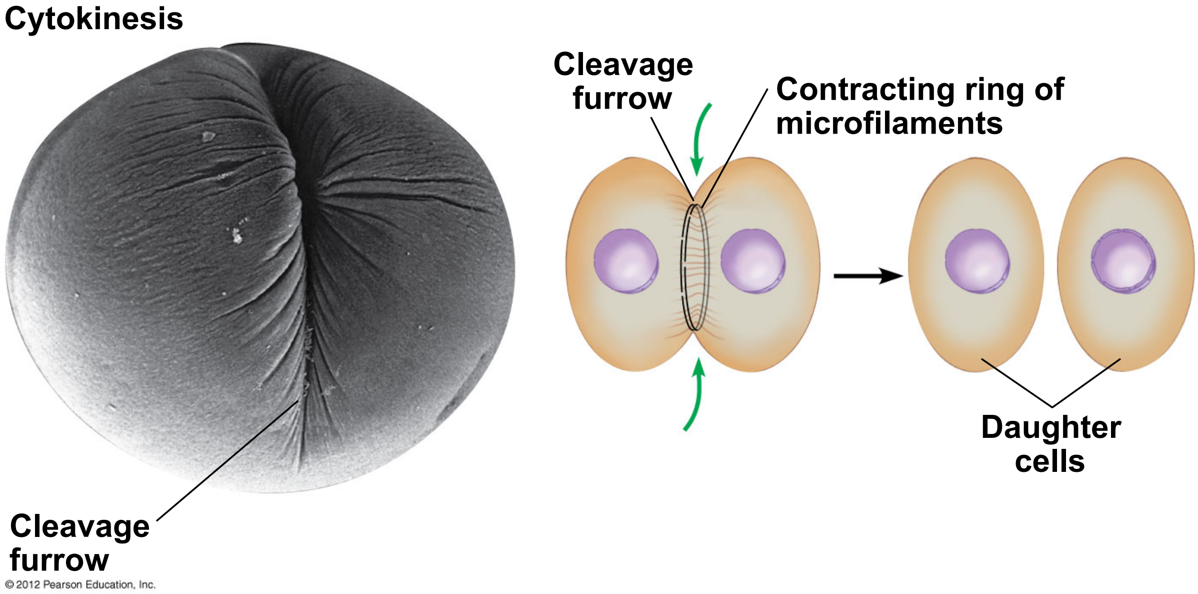
Cell division is a continuum of dynamic changes

- During **cytokinesis**, the cytoplasm is divided into separate cells.
- The process of cytokinesis differs in animal and plant cells.

Cytokinesis differs for plant and animal cells

- In animal cells, cytokinesis occurs as
  1. a **cleavage furrow** forms from a contracting ring of microfilaments, interacting with myosin, and
  2. the cleavage furrow deepens to separate the contents into two cells.

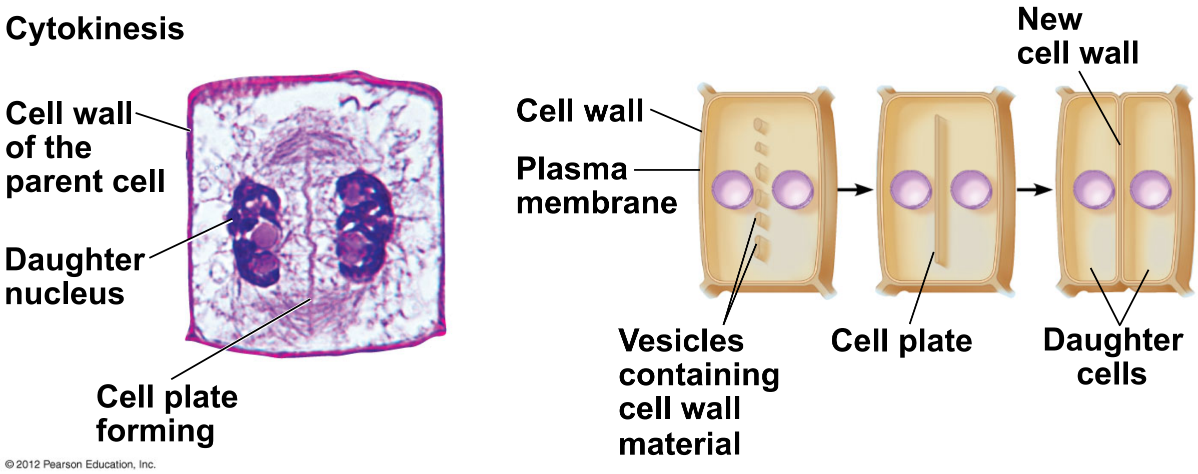
Figure 8.6A



Cytokinesis differs for plant and animal cells

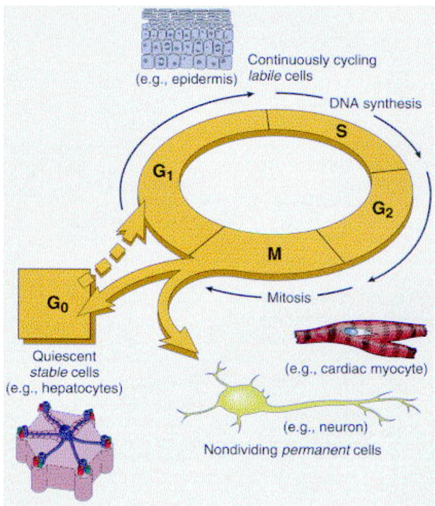
- In plant cells, cytokinesis occurs as
  1. a **cell plate** forms in the middle, from vesicles containing cell wall material,
  2. the cell plate grows outward to reach the edges, dividing the contents into two cells,
  3. each cell now possesses a plasma membrane and cell wall.

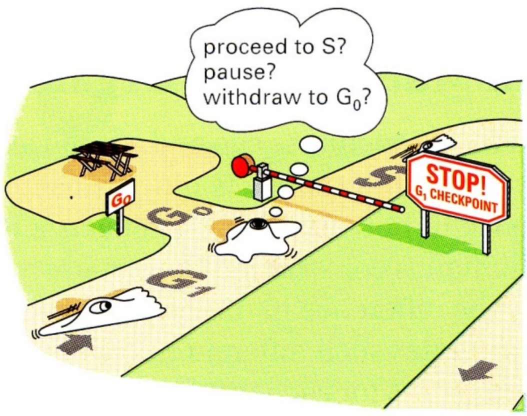
Figure 8.6B



Regulation of Cell Cycle

- Check points or switches control the rate of the cell cycle
- Intracellular and extracellular control
- G0 state is the resting state
- G1 checkpoint or the start checkpoint is said to be the beginning of the cell cycle.





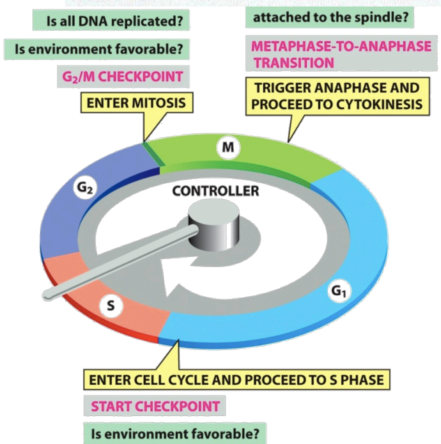


Figure 17-14 Molecular Biology of the Cell 5/e (© Garland Science 2008)

Anchorage, cell density, and chemical growth factors affect cell division

- The cells within an organism's body divide and develop at different rates.
- Cell division is controlled by
  - the presence of essential nutrients,
  - **growth factors**, proteins that stimulate division,
  - **density-dependent inhibition**, in which crowded cells stop dividing, and
  - **anchorage dependence**, the need for cells to be in contact with a solid surface to divide.

Figure 8.7A

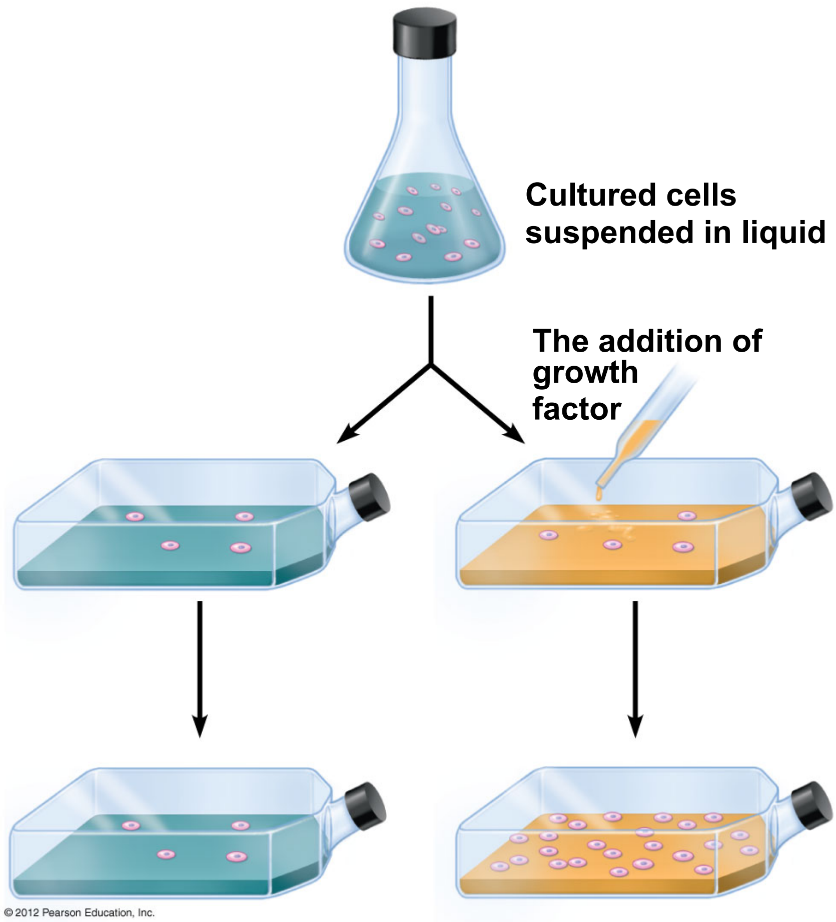
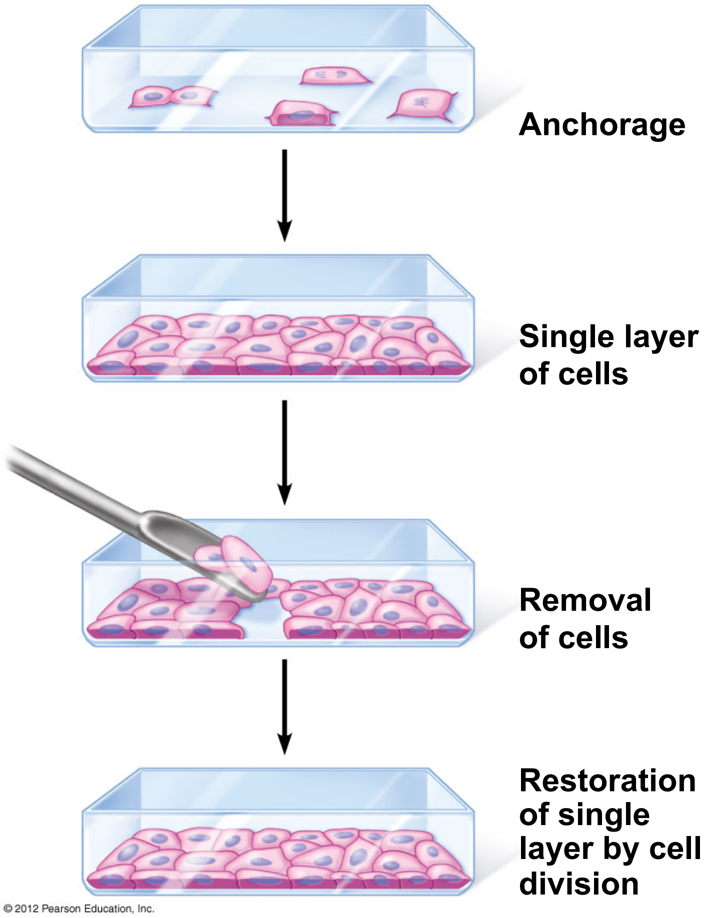


Figure 8.7B



Growth factors signal the cell cycle control system

- The **cell cycle control system** is a cycling set of molecules in the cell that
  - triggers and
  - coordinates key events in the cell cycle.
- Checkpoints in the cell cycle can
  - stop an event or
  - signal an event to proceed.

Growth factors signal the cell cycle control system

- There are three major checkpoints in the cell cycle.
  1. G_1 checkpoint
     - allows entry into the S phase or
     - causes the cell to leave the cycle, entering a nondividing G_0 phase.
  2. G_2 checkpoint, and
  3. M checkpoint.
- Research on the control of the cell cycle is one of the hottest areas in biology today.

Figure 8.8A

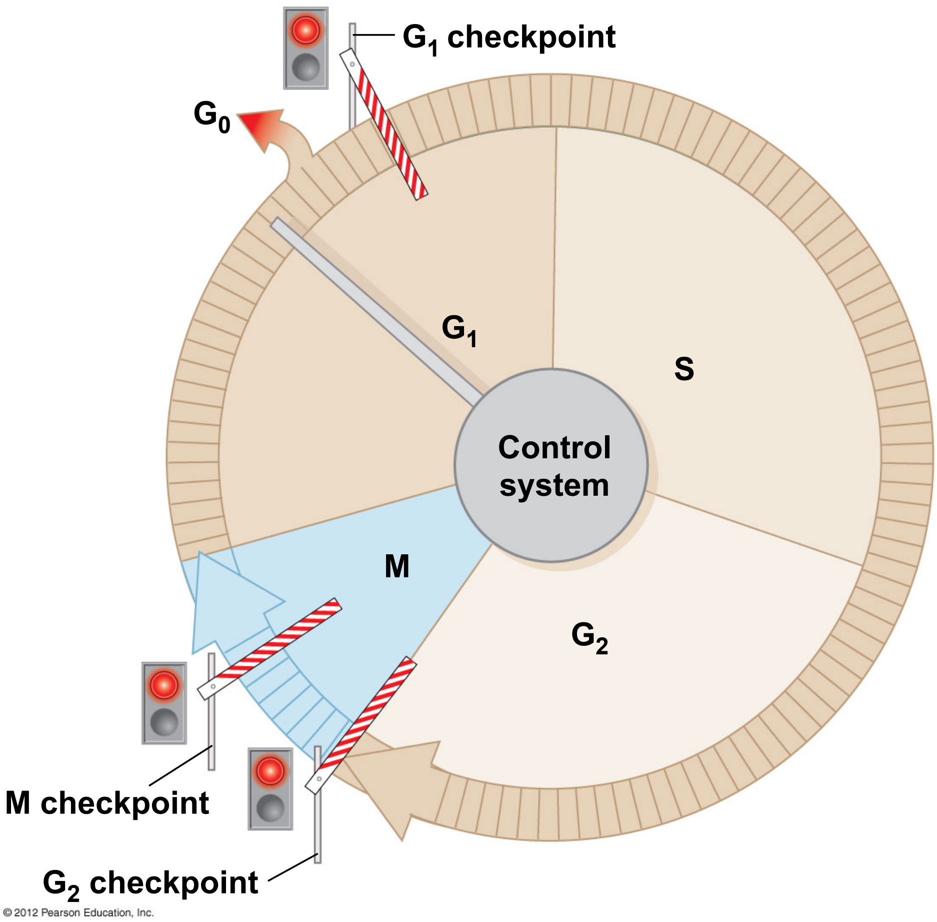
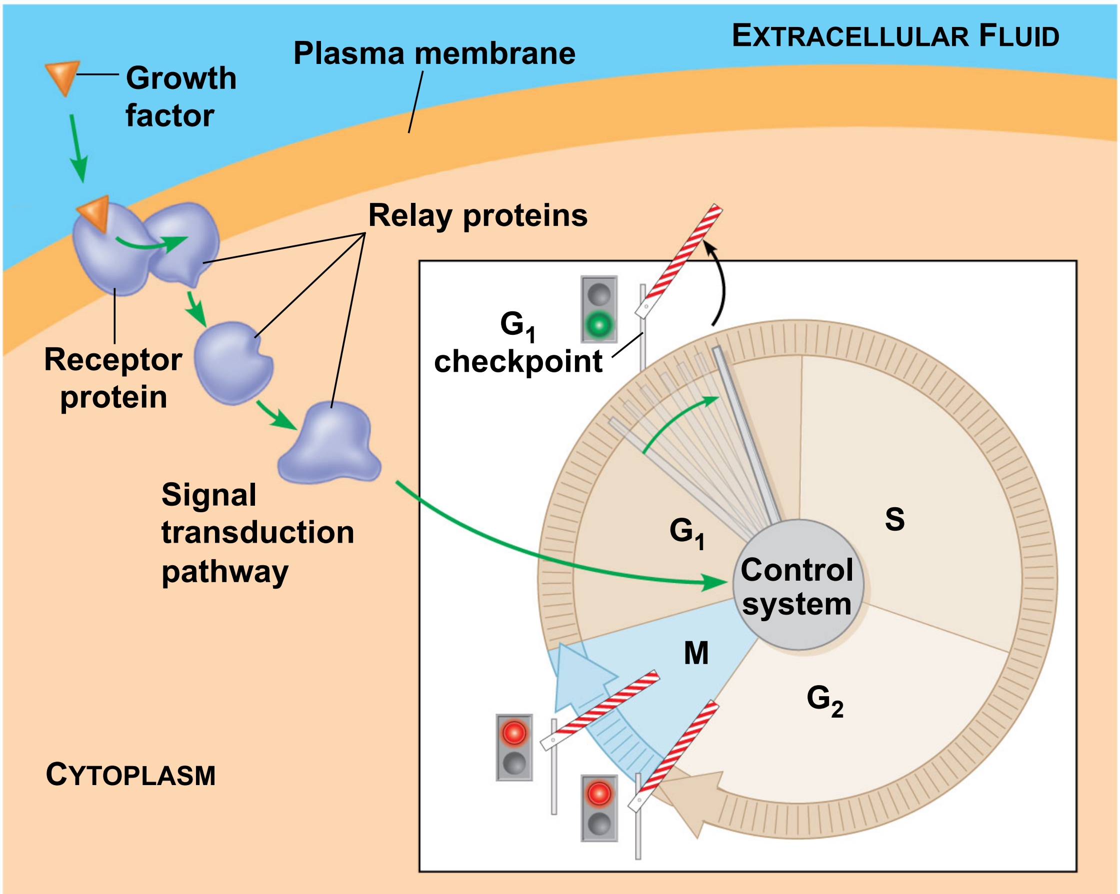


Figure 8.8B

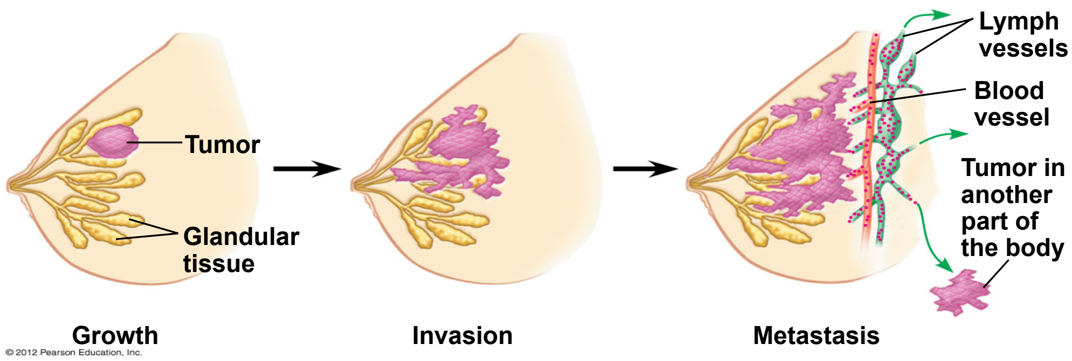


Growing out of control, cancer cells produce malignant tumors

- Cancer currently claims the lives of 20% of the people in the United States and other industrialized nations.
- Cancer cells escape controls on the cell cycle.
- Cancer cells
  - divide rapidly, often in the absence of growth factors,
  - spread to other tissues through the circulatory system, and
  - grow without being inhibited by other cells.

CONNECTION: Growing out of control, cancer cells produce malignant tumors

- A **tumor** is an abnormally growing mass of body cells.
  - **Benign tumors** remain at the original site.
  - **Malignant tumors** spread to other locations, called **metastasis**.

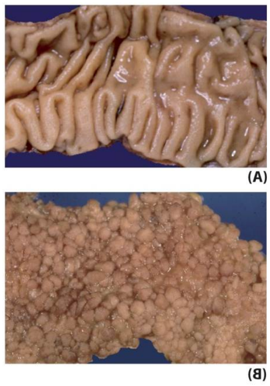


Growing out of control, cancer cells produce malignant tumors

- Cancers are named according to the organ or tissue in which they originate.
  - **Carcinomas** arise in external or internal body coverings.
  - **Sarcomas** arise in supportive and connective tissue.
  - **Leukemias** and **lymphomas** arise from blood-forming tissues.

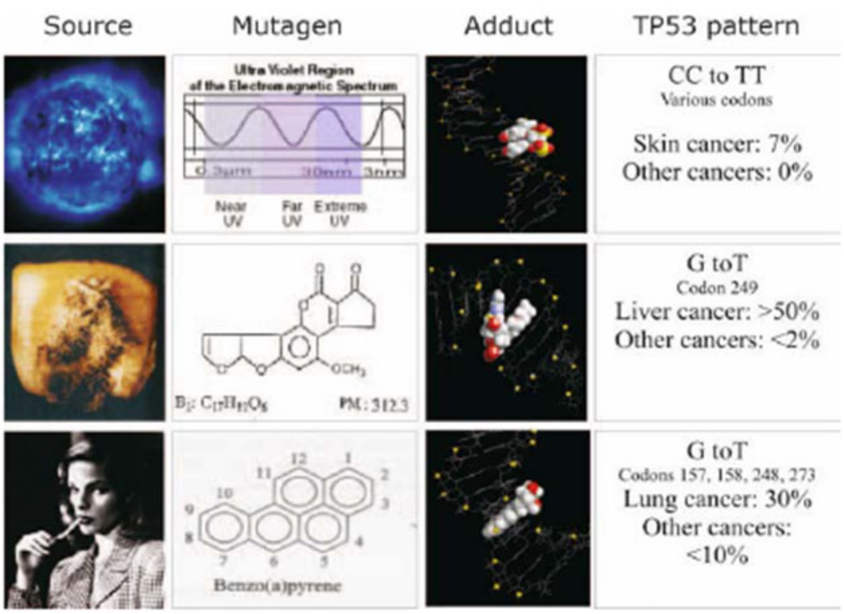
Growing out of control, cancer cells produce malignant tumors

- Cancer treatments
  - Localized tumors can be
    - removed surgically and/or
    - treated with concentrated beams of high-energy radiation.
  - Chemotherapy is used for metastatic tumors.



Tumor suppressor mutation and cancer

- ATM/ATR (Ataxia telangiectasia)
- P53 (tumor suppressor gene) regulate cell cycle



MEIOSIS AND CROSSING OVER

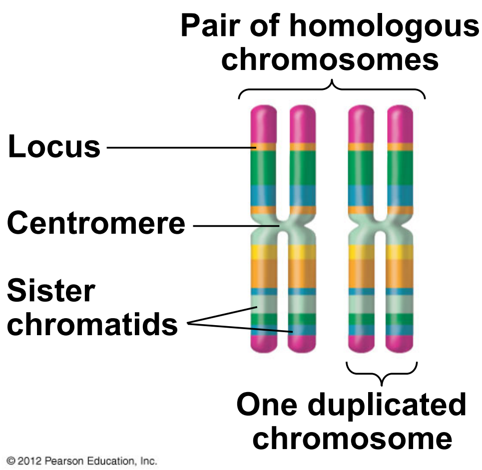
Chromosomes are matched in homologous pairs

- In humans, **somatic cells** have
  - 23 pairs of homologous chromosomes and
  - one member of each pair from each parent.
- The human **sex chromosomes** X and Y differ in size and genetic composition.
- The other 22 pairs of chromosomes are **autosomes** with the same size and genetic composition.

Chromosomes are matched in homologous pairs

- **Homologous chromosomes** are matched in
  - length,
  - centromere position, and
  - gene locations.
- A **locus** (plural, *loci*) is the position of a gene.
- Different versions of a gene may be found at the same locus on maternal and paternal chromosomes.

Figure 8.11



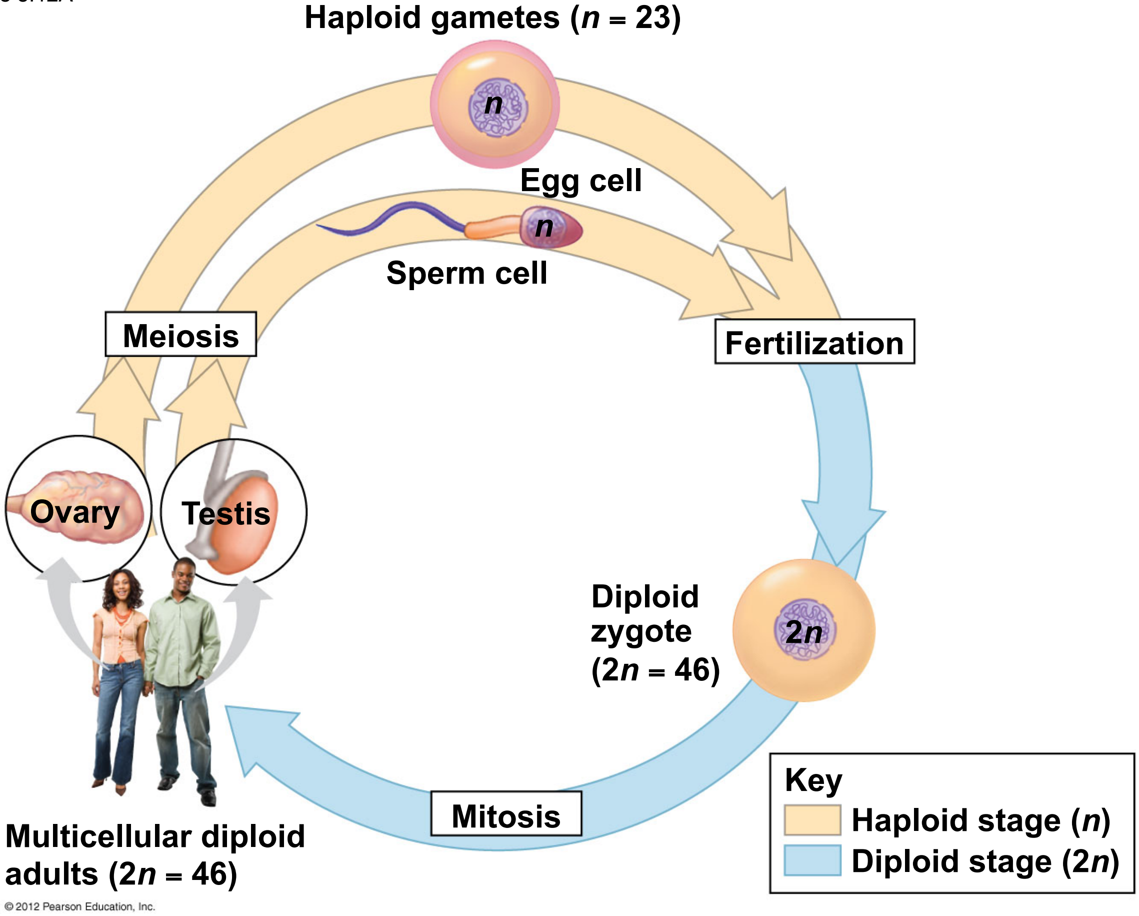
Gametes have a single set of chromosomes

- An organism's **life cycle** is the sequence of stages leading
  - from the adults of one generation
  - to the adults of the next.
- Humans and many animals and plants are diploid, with body cells that have
  - two sets of chromosomes,
  - one from each parent.

Gametes have a single set of chromosomes

- Meiosis is a process that converts diploid nuclei to haploid nuclei.
  - **Diploid cells** have two homologous sets of chromosomes.
  - **Haploid cells** have one set of chromosomes.
  - Meiosis occurs in the sex organs, producing **gametes** —sperm and eggs.
- **Fertilization** is the union of sperm and egg.
- The **zygote** has a diploid chromosome number, one set from each parent.

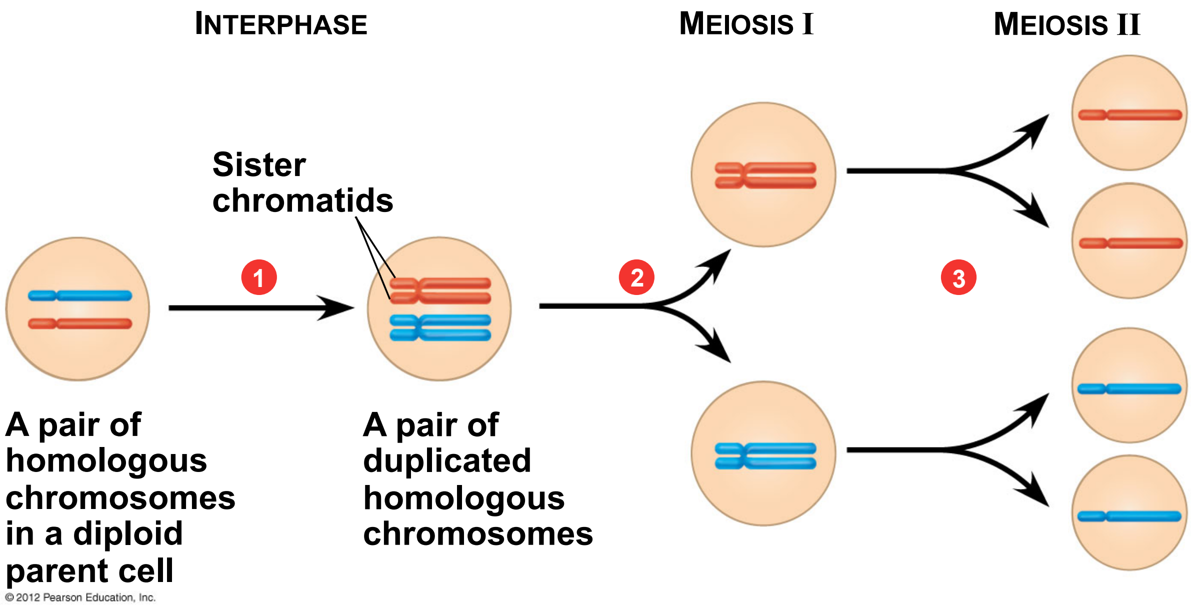
Figure 8.12A



Gametes have a single set of chromosomes

- All sexual life cycles include an alternation between
  - a diploid stage and
  - a haploid stage.
- Producing haploid gametes prevents the chromosome number from doubling in every generation.

Figure 8.12B



Meiosis reduces the chromosome number from diploid to haploid

- **Meiosis** is a type of cell division that produces haploid gametes in diploid organisms.
- Two haploid gametes combine in fertilization to restore the diploid state in the zygote.

Meiosis reduces the chromosome number from diploid to haploid

- Meiosis and mitosis are preceded by the duplication of chromosomes. However,
  - meiosis is followed by two consecutive cell divisions and
  - mitosis is followed by only one cell division.
- Because in meiosis, one duplication of chromosomes is followed by two divisions, each of the four daughter cells produced has a haploid set of chromosomes.

Meiosis reduces the chromosome number from diploid to haploid

- **Meiosis I** – **Prophase I** – events occurring in the nucleus.
  - Chromosomes coil and become compact.
  - Homologous chromosomes come together as pairs by **synapsis**.
  - Each pair, with four chromatids, is called a tetrad.
  - Nonsister chromatids exchange genetic material by crossing over.

Figure 8.13_left

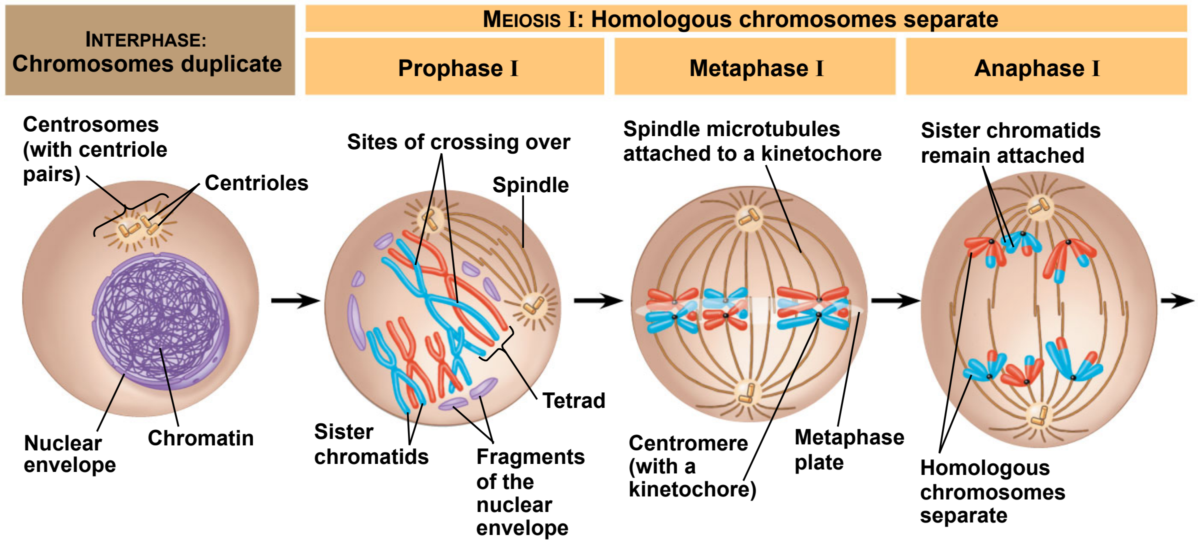


Figure 8.13_right

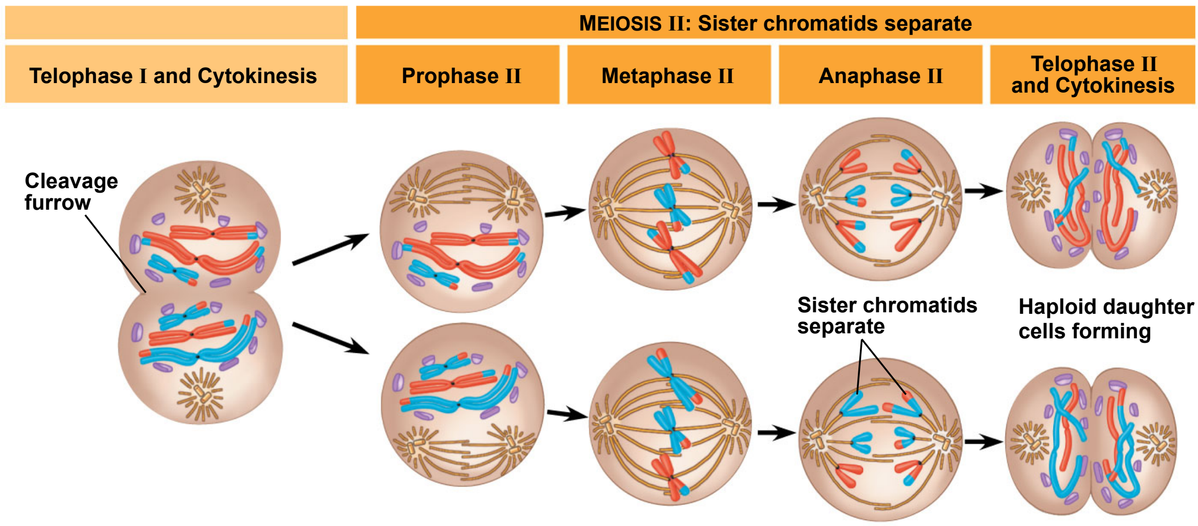
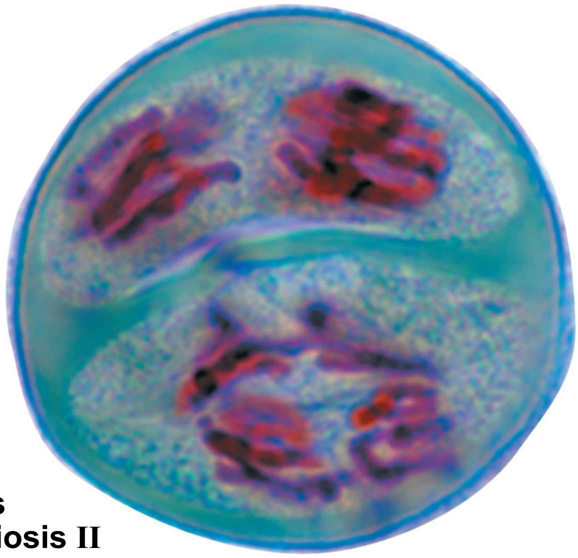


Figure 8.13_5



Two lily cells undergo meiosis II

Mitosis and meiosis have important similarities and differences

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

Figure 8.14

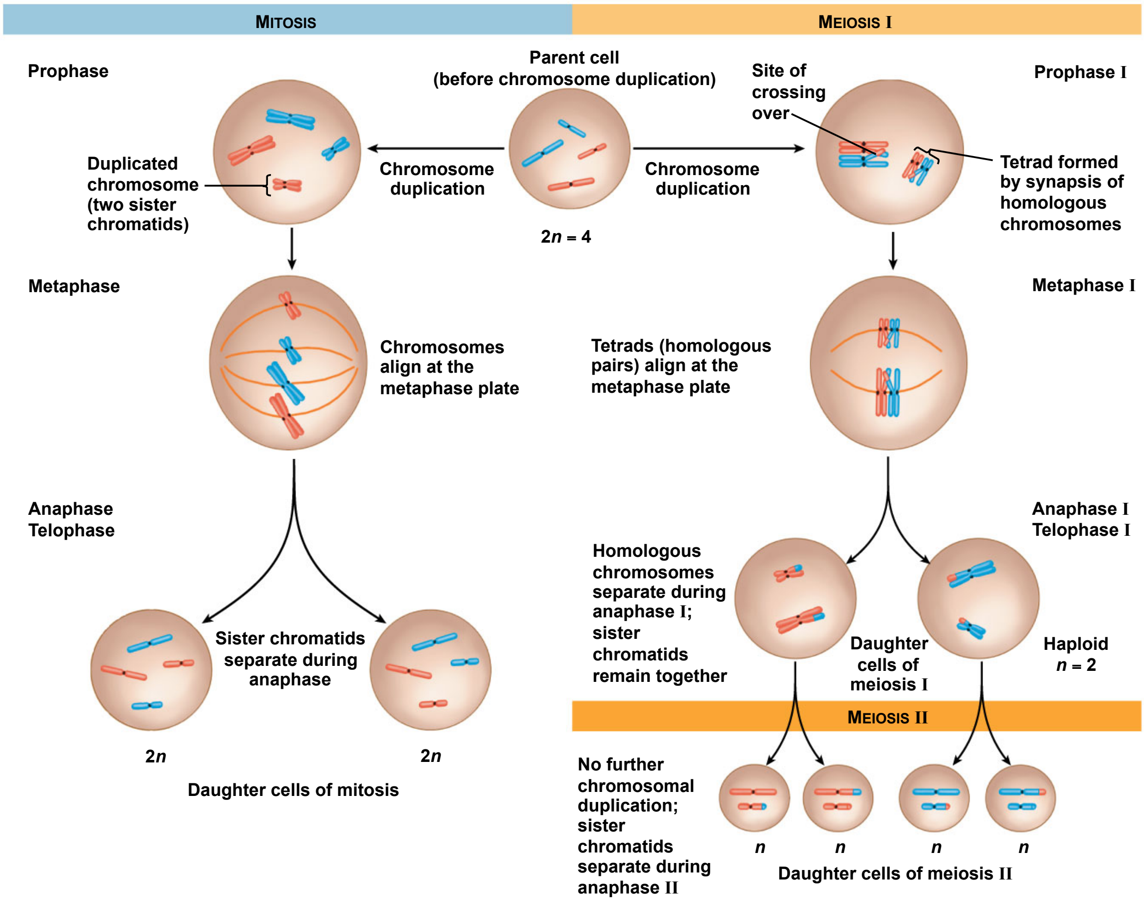


Figure 8.14_1

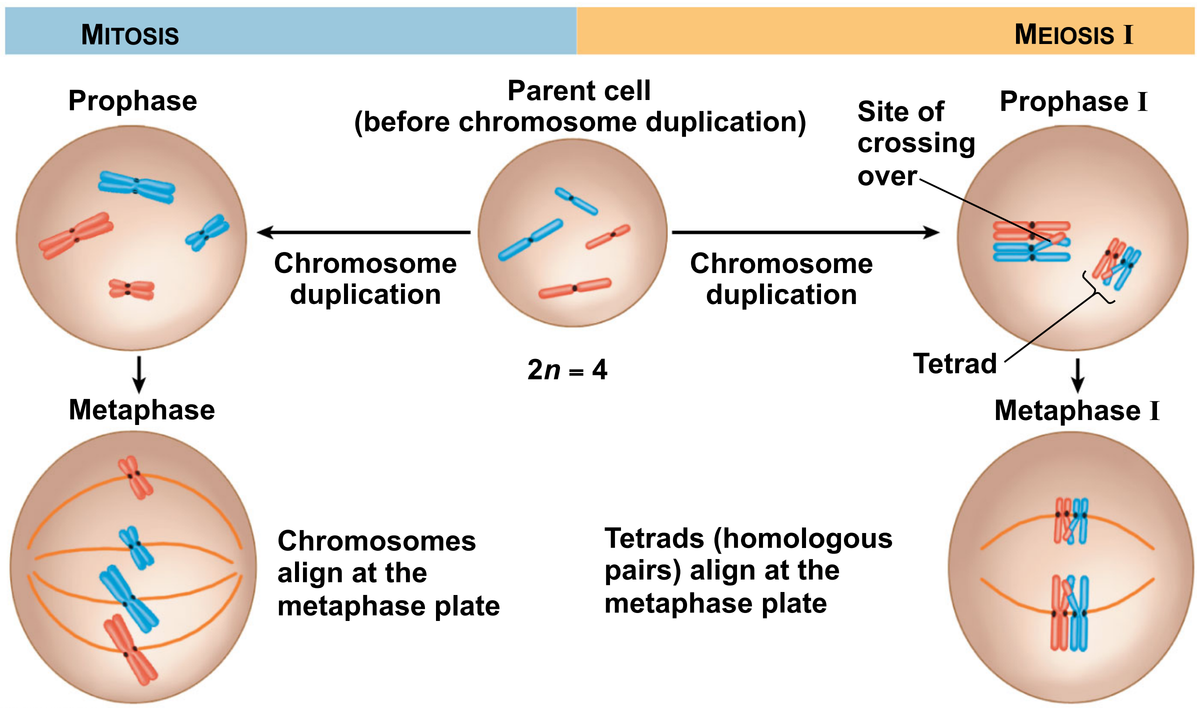


Figure 8.14_2

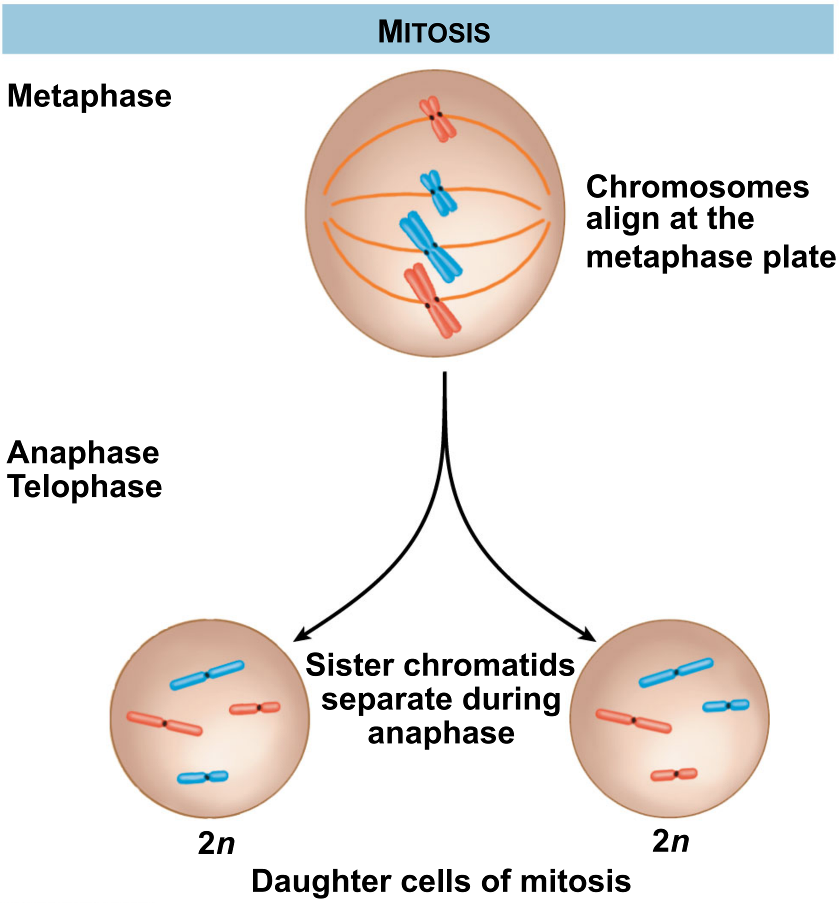
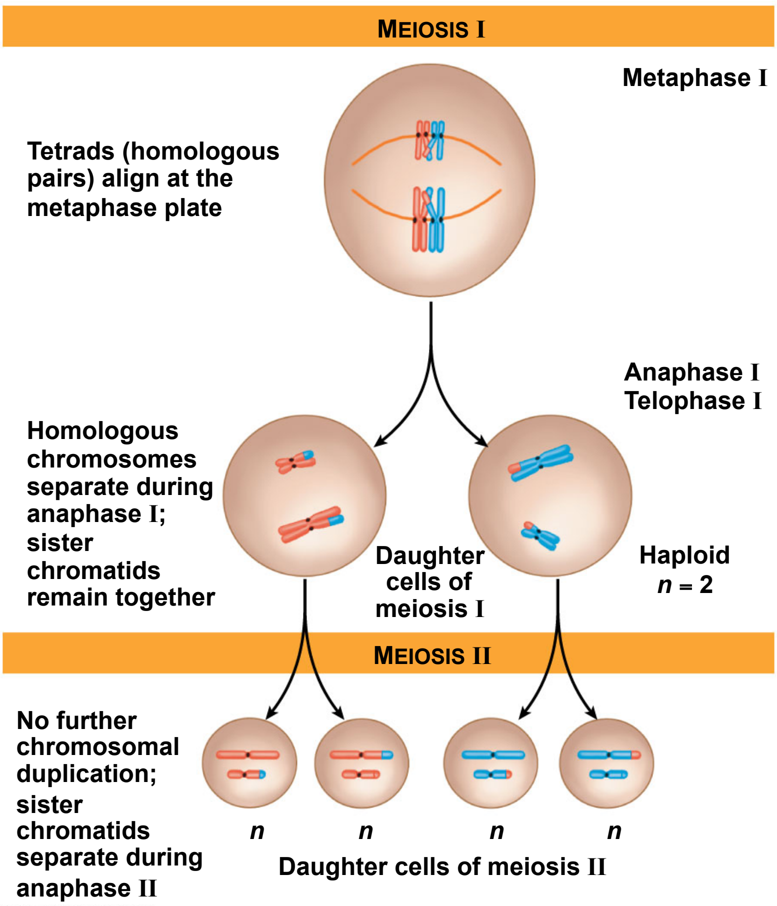


Figure 8.14_3



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

- Genetic variation in gametes results from
  – independent orientation at metaphase I and
  – random fertilization.

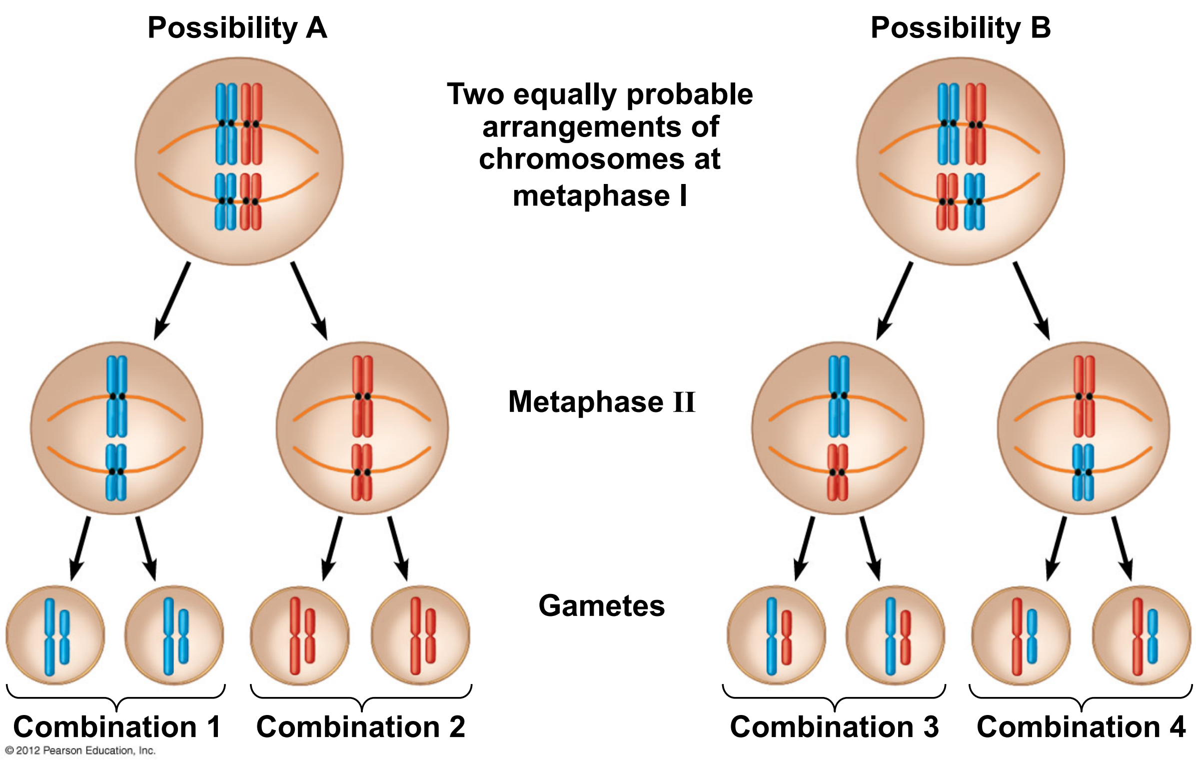
Independent orientation of chromosomes in meiosis and random fertilization lead to varied offspring

- Independent orientation at metaphase I
  – Each pair of chromosomes independently aligns at the cell equator.
  – There is an equal probability of the maternal or paternal chromosome facing a given pole.
  – The number of combinations for chromosomes packaged into gametes is 2^n where n = haploid number of chromosomes.

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

- Random fertilization – The combination of each unique sperm with each unique egg increases genetic variability.

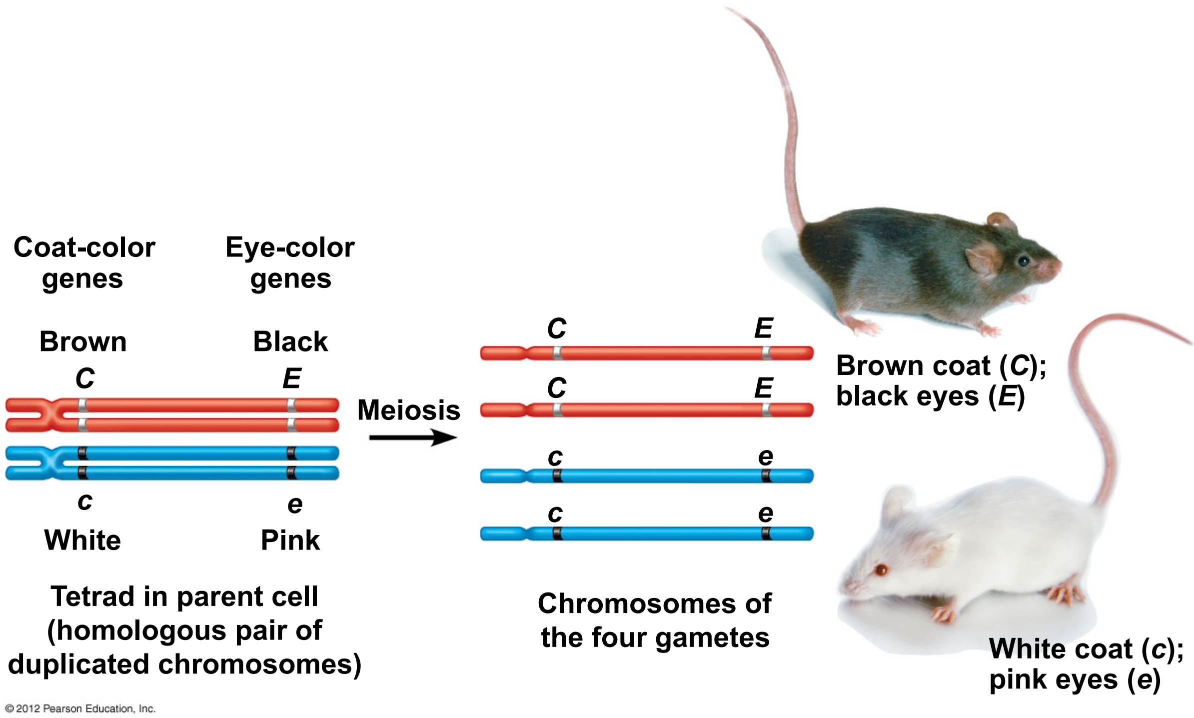
Figure 8.15_s3



Homologous chromosomes may carry different versions of genes

- Separation of homologous chromosomes during meiosis can lead to genetic differences between gametes.
  - Homologous chromosomes may have different versions of a gene at the same locus.
  - One version was inherited from the maternal parent and the other came from the paternal parent.
  - Since homologues move to opposite poles during anaphase I, gametes will receive either the maternal or paternal version of the gene.

Figure 8.16



Crossing over further increases genetic variability

- **Genetic recombination** is the production of new combinations of genes due to crossing over.
- **Crossing over** is an exchange of corresponding segments between separate (nonsister) chromatids on homologous chromosomes.
  - Nonsister chromatids join at a **chiasma** (plural, *chiasmata*), the site of attachment and crossing over.
  - Corresponding amounts of genetic material are exchanged between maternal and paternal (nonsister) chromatids.

Figure 8.17B_1

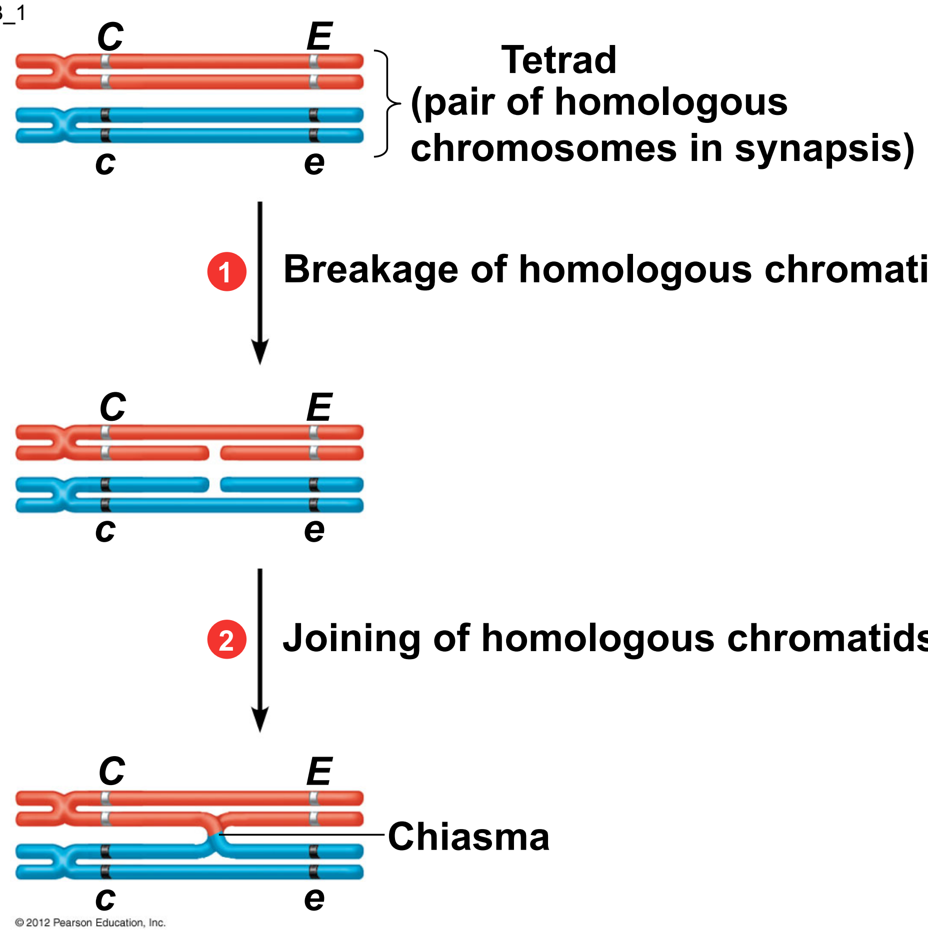


Figure 8.17B_2

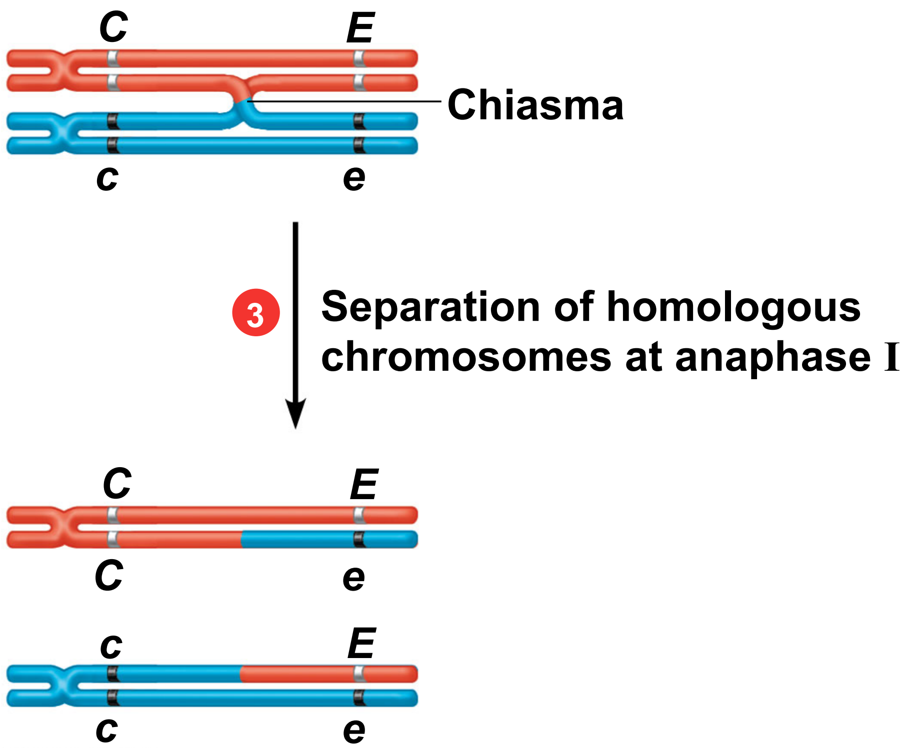
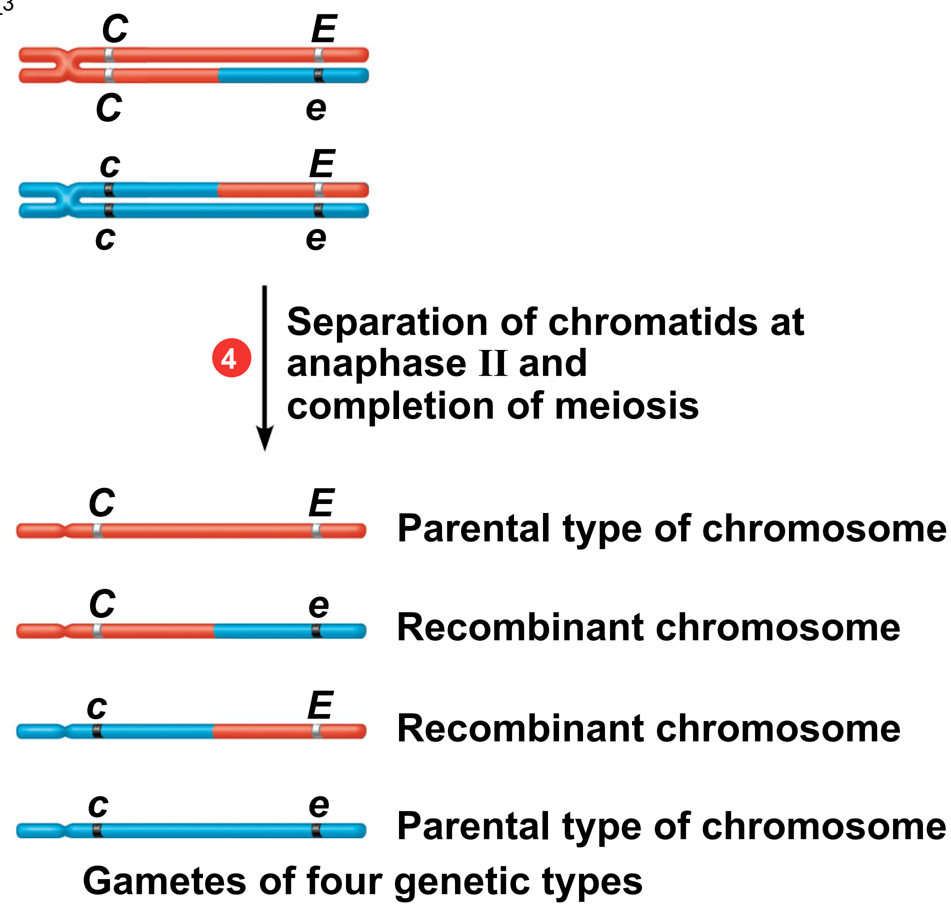


Figure 8.17B_3



ALTERATIONS OF CHROMOSOME NUMBER AND STRUCTURE

A karyotype is a photographic inventory of an individual’s chromosomes

- A **karyotype** is an ordered display of magnified images of an individual’s chromosomes arranged in pairs.
- Karyotypes
  - are often produced from dividing cells arrested at metaphase of mitosis and
  - allow for the observation of
    - homologous chromosome pairs,
    - chromosome number, and
    - chromosome structure.

– Preparation of a karyotype from a blood sample

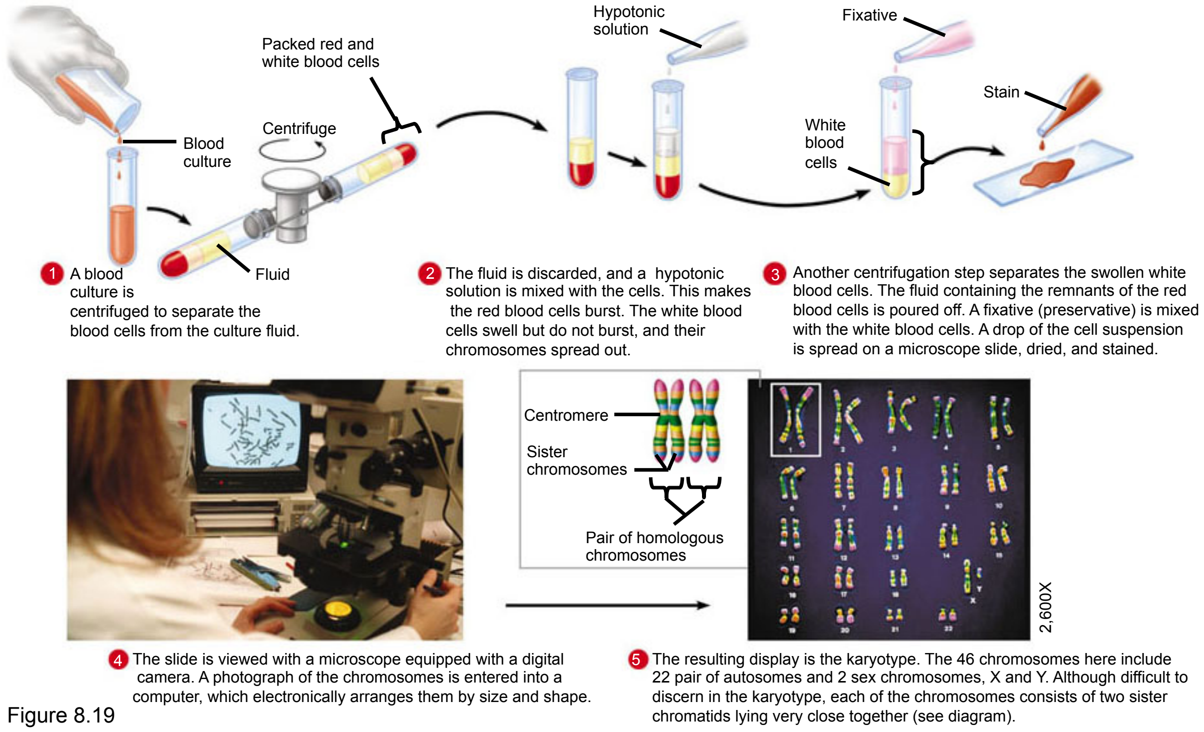
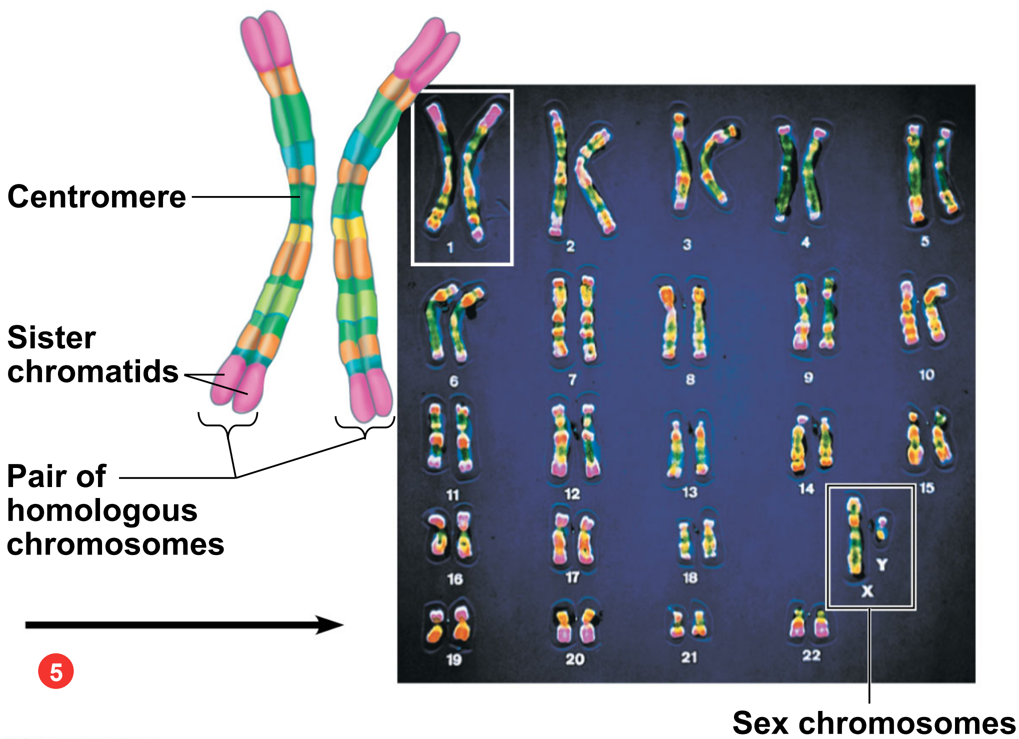


Figure 8.19

Figure 8.18_s5

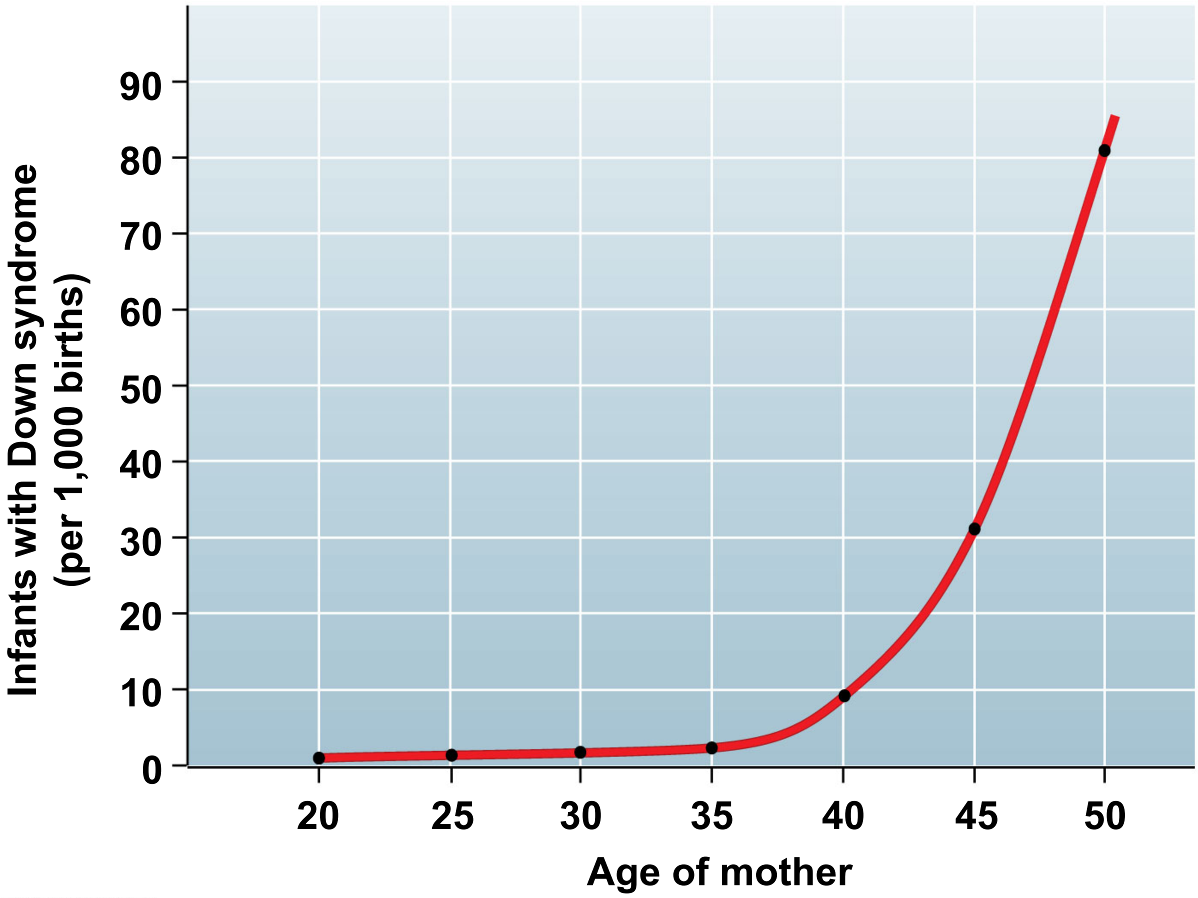


Case : Down syndrome - an extra copy of chromosome 21

- **Trisomy 21 – Down syndrome**
  - involves the inheritance of three copies of chromosome 21 and
  - is the most common human chromosome abnormality.
- symptoms, :
  - mental retardation,
  - characteristic facial features,
  - short stature,
  - heart defects,
  - susceptibility to respiratory infections, leukemia, and Alzheimer’s disease, and
  - shortened life span.
- The incidence increases with the age of the mother.

Figure 8.19A

Figure 8.19B



Accidents during meiosis can alter chromosome number

- **Nondisjunction** is the failure of chromosomes or chromatids to separate normally during meiosis. This can happen during
  - meiosis I, if both members of a homologous pair go to one pole or
  - meiosis II if both sister chromatids go to one pole.
- Fertilization after nondisjunction yields zygotes with altered numbers of chromosomes.

Figure 8.20A_s3

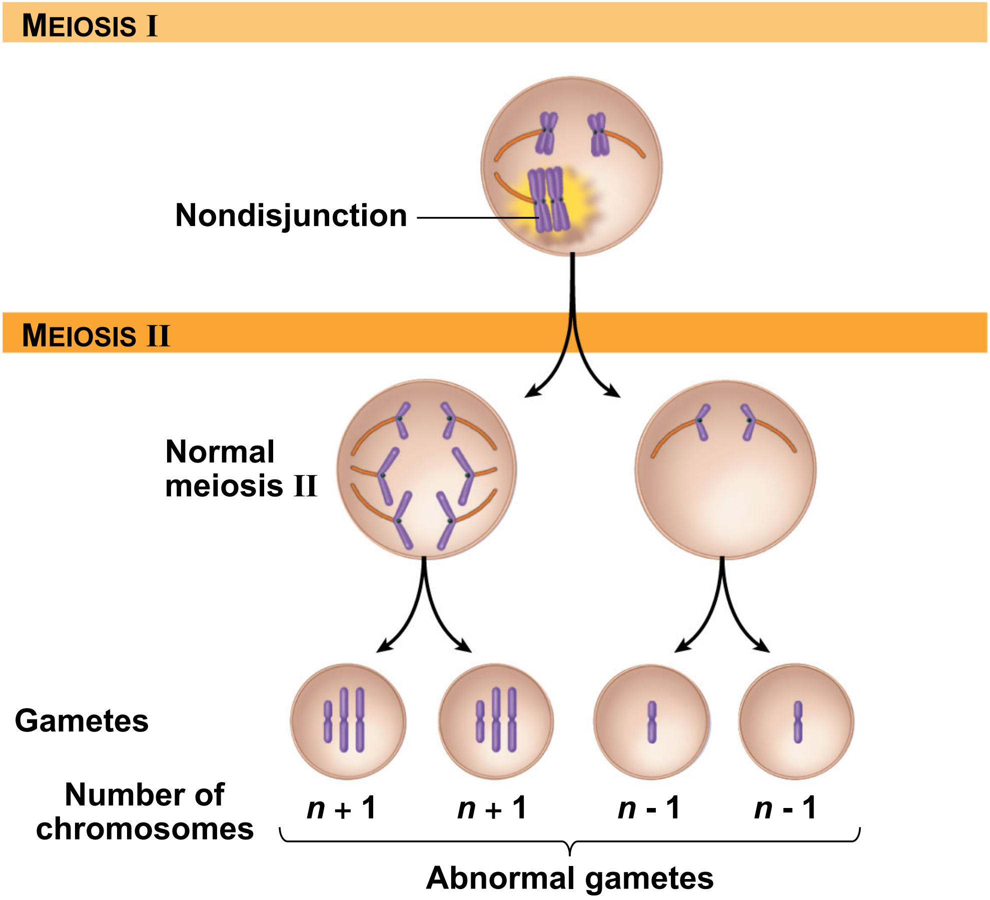
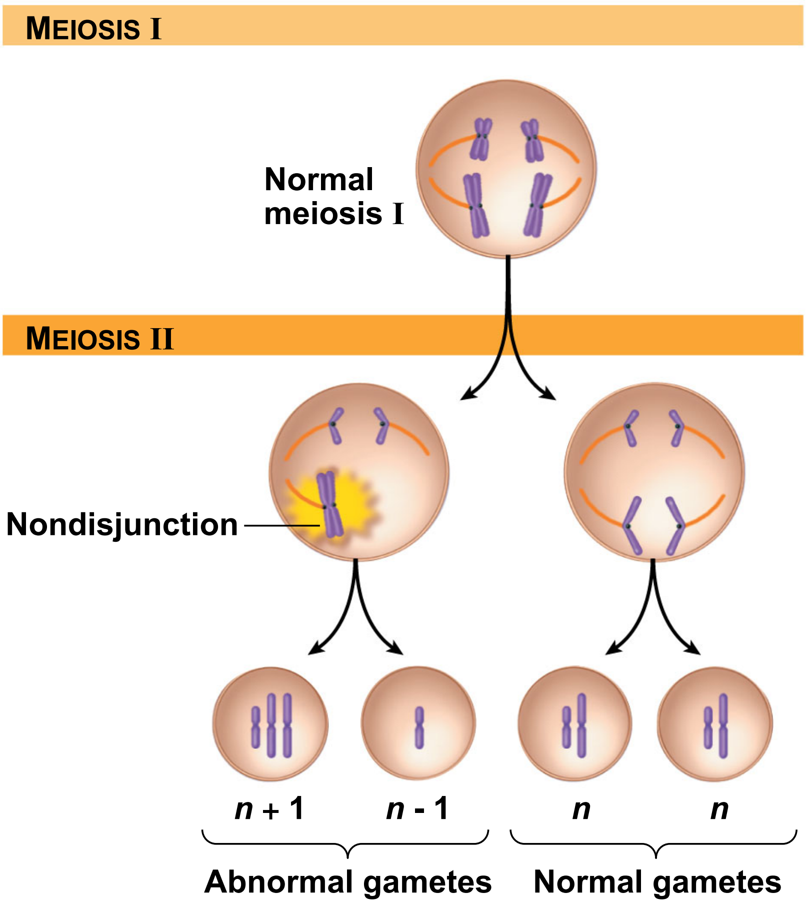
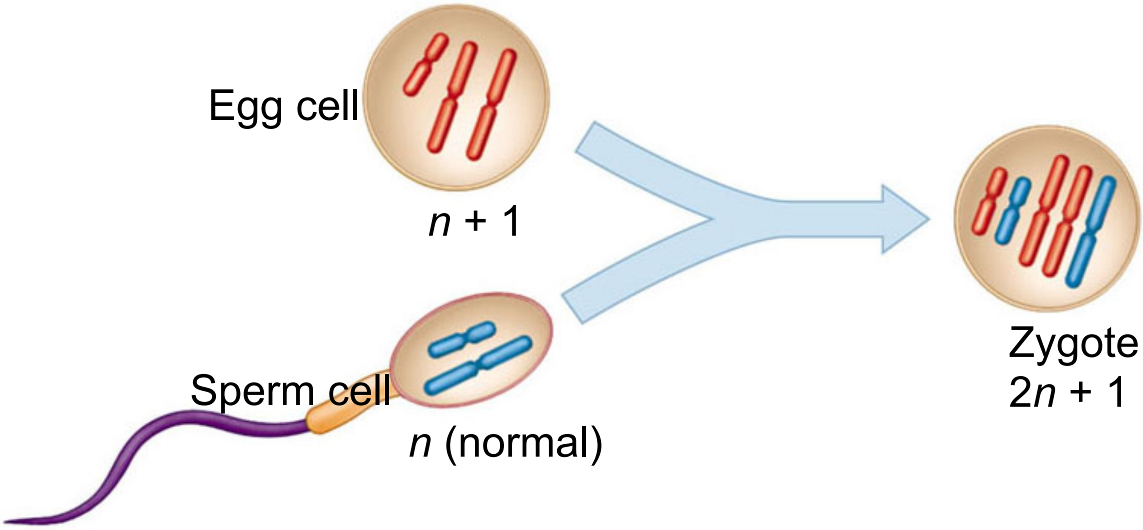


Figure 8.20B_s3



Abnormal numbers of sex chromosomes do not usually affect survival

Nondisjunction can also produce gametes with extra or missing sex chromosomes



– Fertilization after nondisjunction in the mother

Figure 8.21C

– Human sex chromosome abnormalities

TABLE 8.22 ABNORMALITIES OF SEX CHROMOSOME NUMBER IN HUMANS

Sex Chromosomes	Syndrome	Origin of Nondisjunction	Frequency in Population
XXY	Klinefelter syndrome (male)	Meiosis in egg or sperm formation	$\frac{1}{2,000}$
XYY	None (normal male)	Meiosis in sperm formation	$\frac{1}{2,000}$
XXX	None (normal female)	Meiosis in egg or sperm formation	$\frac{1}{1,000}$
XO	Turner syndrome (female)	Meiosis in egg or sperm formation	$\frac{1}{5,000}$

CONNECTION

- 8.23 Alterations of chromosome structure can cause birth defects and cancer
  - Chromosome breakage can lead to rearrangements
    - That can produce genetic disorders or, if the changes occur in somatic cells, cancer

Abnormal numbers of sex chromosomes do not usually affect survival

- Sex chromosome abnormalities tend to be less severe, perhaps because of
  - the small size of the Y chromosome or
  - X-chromosome inactivation.

Alterations of chromosome structure can cause birth defects and cancer

- Chromosome breakage can lead to rearrangements that can produce
  - genetic disorders or,
  - if changes occur in somatic cells, cancer.

Alterations of chromosome structure can cause birth defects and cancer

- These rearrangements may include
  - **a deletion**, the loss of a chromosome segment,
  - **a duplication**, the repeat of a chromosome segment,
  - **an inversion**, the reversal of a chromosome segment, or
  - **a translocation**, the attachment of a segment to a nonhomologous chromosome that can be reciprocal.

Alterations of chromosome structure can cause birth defects and cancer

- Chronic myelogenous leukemia (CML)
  - is one of the most common leukemias,
  - affects cells that give rise to white blood cells (leukocytes), and
  - results from part of chromosome 22 switching places with a small fragment from a tip of chromosome 9.

Figure 8.23A

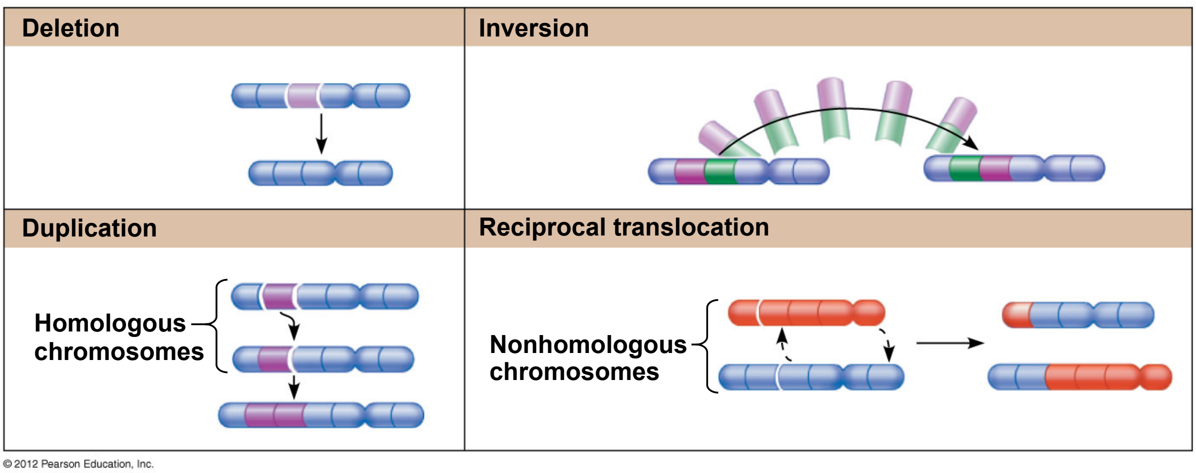
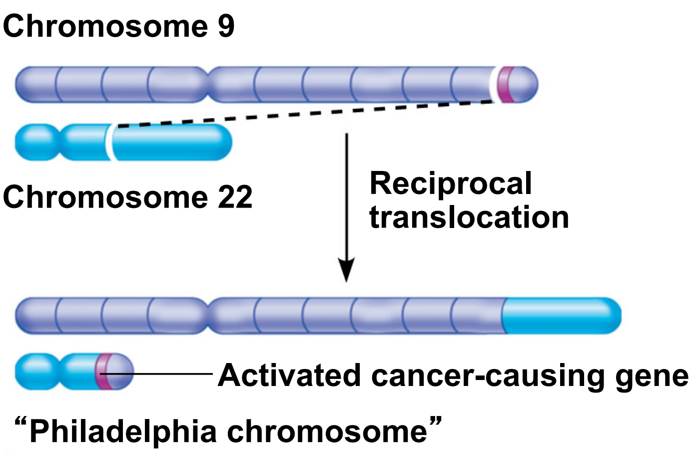


Figure 8.23B


Chromosome 9
Chromosome 22
Reciprocal translocation
Activated cancer-causing gene
"Philadelphia chromosome"