

Essentials of Biology

Sylvia S. Mader

Chapter 9
Meiosis and Down Syndrome
Lecture Outline

Children with Down Syndrome



Down Syndrome

- Trisomy 21
- Short stature, eyelid fold, stubby fingers, mental disabilities
- High rate of ..
 - ✓ Alzheimer's
 - ✓ Heart defects
 - ✓ Hearing loss
 - ✓ Vision problems
- Chance of a woman having a Down syndrome child increases rapidly with age

Is this a normal Karyotype?

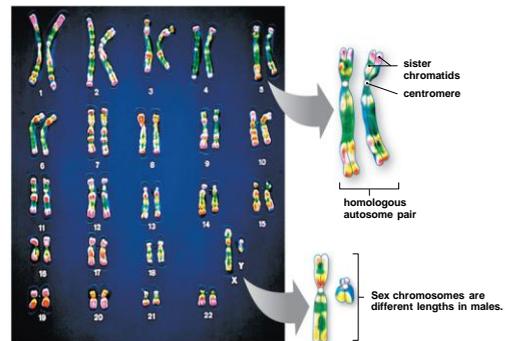


Figure 9.1

What are homologous chromosomes & where do they come from?

- One member of each homologous pair from each parent.
- Homologous pairs may contain different versions of the same gene.
 - **Alleles** – alternate forms of a gene! Examples??
- Both males and females have 23 pairs of chromosomes.
- 23 pairs or 46 total chromosomes = **diploid number (2n)**
 - **Somatic Cells** have the diploid number of chromosomes
- **Haploid number (n) in gametes** = 23 total chromosomes
 - 22 pairs of **autosomes**
 - 1 pair of **sex chromosomes**
 - XX female or XY male

Down Syndrome Karyotype

- Down syndrome due to an error in meiosis
 - What's meiosis?
- What is wrong with the Karyotype?
- Why are most trisomies fatal?
- Trisomies involving the sex chromo's sex chromosomes are not fatal.

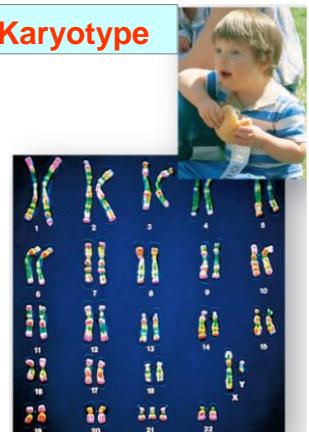
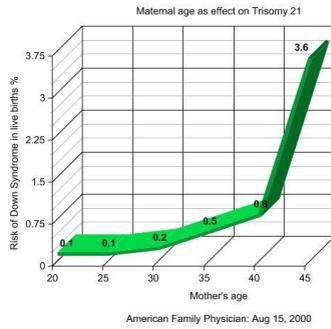


Figure 9.10

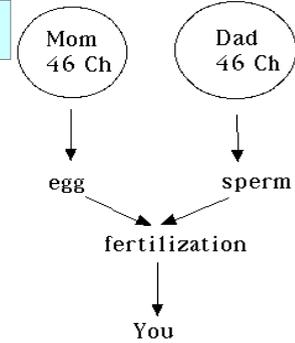
Down Syndrome—As a function of mother's age

- 1 in 1000 births in U.S.
- 1 in 12 births at age 50
- Most frequent genetic cause of mental retardation
 - I.Q. = 20-50



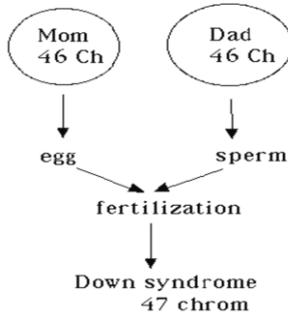
Human Somatic Cells have 46 Chromosomes

- How many chromosomes in human gametes?
- How do you know?



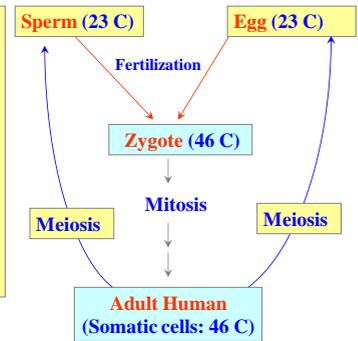
Down Syndrome

- How many chromosomes are in the egg?
- Sperm?
- How do you know?
- D.S. is due to an error in Meiosis



Human Life Cycle

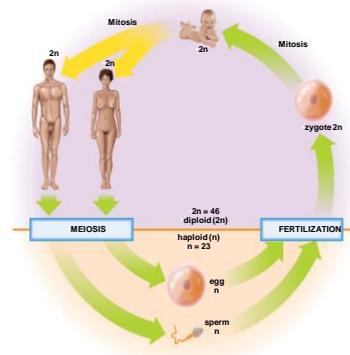
1. Role of mitosis?
 - produces gametes
 - A reductive division (46 → 23 chromosomes)
 - Don't confuse meiosis with mitosis
2. Meiosis
3. What if gametes were made by mitosis?



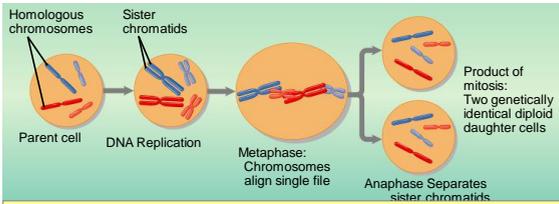
Human life cycle

- **Life cycle**
 - in sexually reproducing organisms refers to all the reproductive events that occur from one generation to the next
- Involves both mitosis and meiosis
- **Mitosis involved in continued growth of child and repair of tissues throughout life**
 - Somatic (body) cells are diploid
- **Meiosis reduces the chromosome number from diploid to haploid.**
 - Gametes (egg and sperm) have only 1 member of each homologous pair.
 - Spermatogenesis produces sperm in the testes.
 - Oogenesis produces eggs in the ovaries.
- **Fertilization:** Egg and sperm join to form diploid zygote.

Figure 9.2 Life cycle of humans



Review of Mitosis



1. How do chromo's align at metaphase?
2. What separates at anaphase?
3. How do the daughter cells compare genetically?

9.2 The Phases of Meiosis

- Meiosis involves two divisions: meiosis I and meiosis II.
 - Each division is broken down into four phases:
 - Prophase (I and II)
 - Metaphase (I and II)
 - Anaphase (I and II)
 - Telophase (I and II)

Figure 9.6 Meiosis I

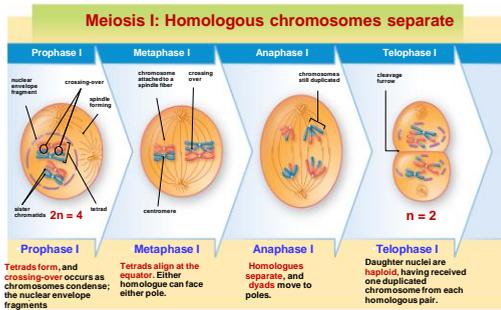
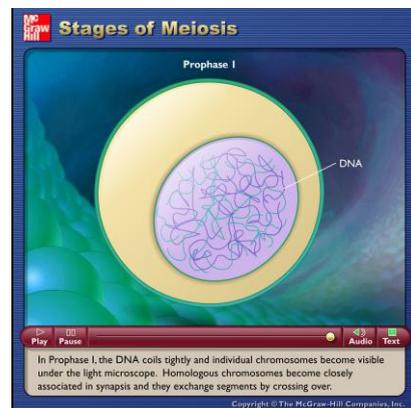
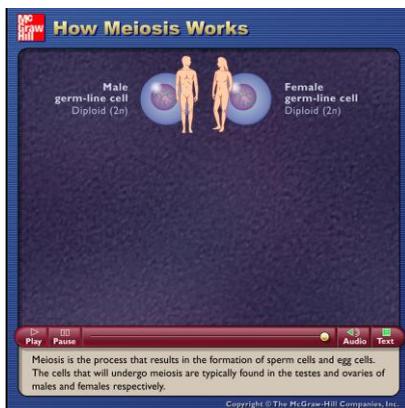
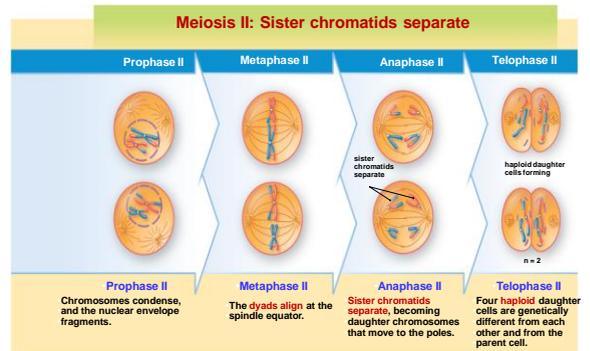
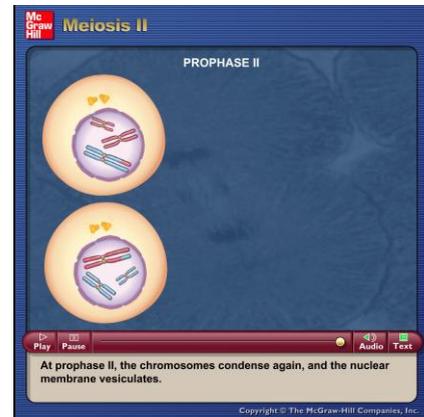
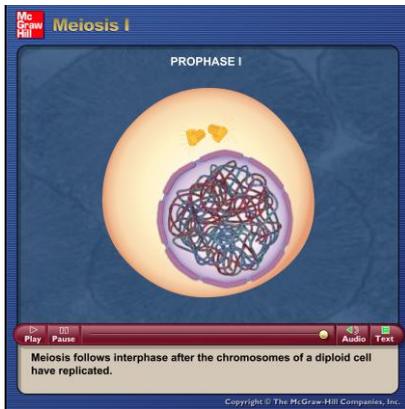


Figure 9.7 Meiosis II





9.4 Abnormal Chromosome Inheritance

- **Nondisjunction**
 - Meiosis I when both members of a pair go into the same daughter cell
 - Meiosis II when sister chromatids fail to separate
- **Trisomy** – 3 copies of a chromosome
- **Monosomy** – single copy of a chromosome

Figure 9.9a Nondisjunction during meiosis I

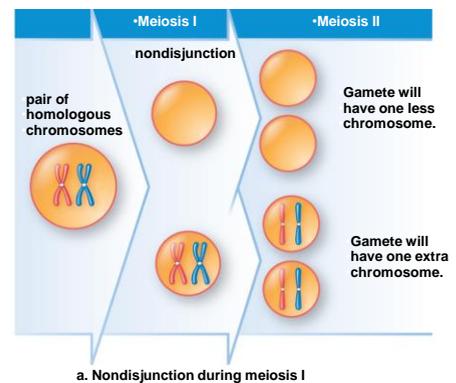
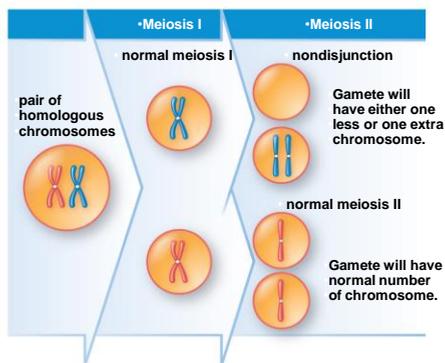
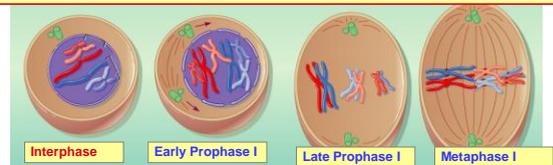


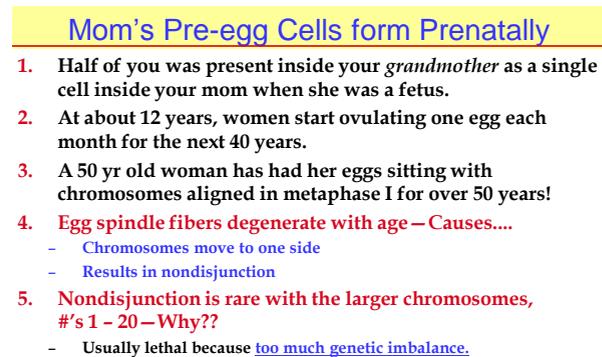
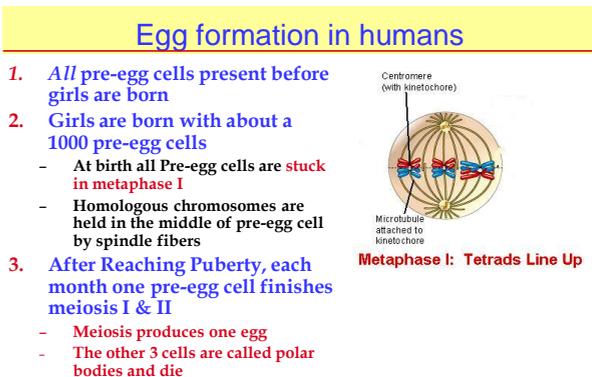
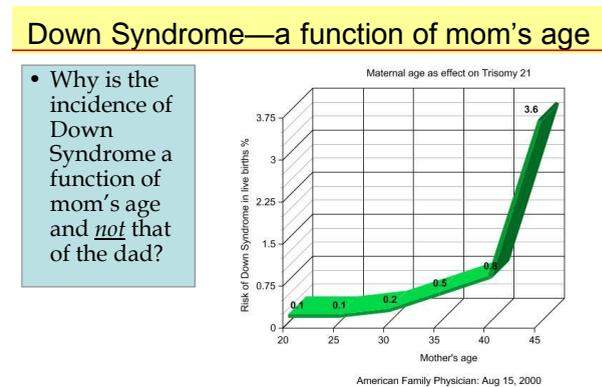
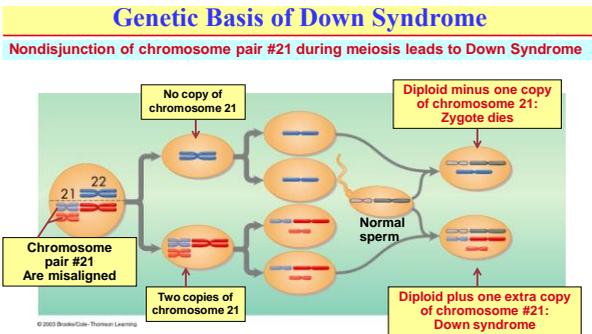
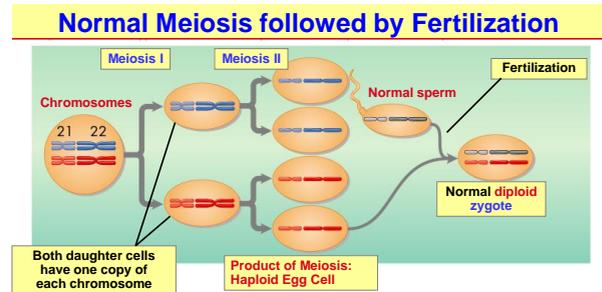
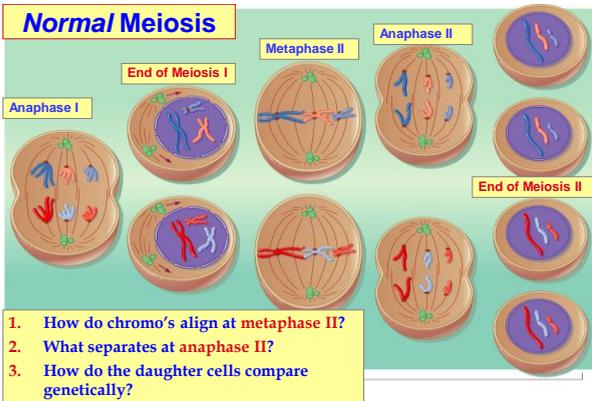
Figure 9.9b Nondisjunction during meiosis II



Normal Meiosis



1. How do chromo's align at metaphase I?
2. What separates at anaphase I?
3. How do the daughter cells compare genetically?



Why doesn't the age of the father influence the incidence of Down syndrome?

1. Sex cell formation in Males

- Sperm formation starts at puberty and continues daily for life
- Each pre-sperm cell divides twice to produce 4 sperm
 - 200-300 million sperm produced per day!

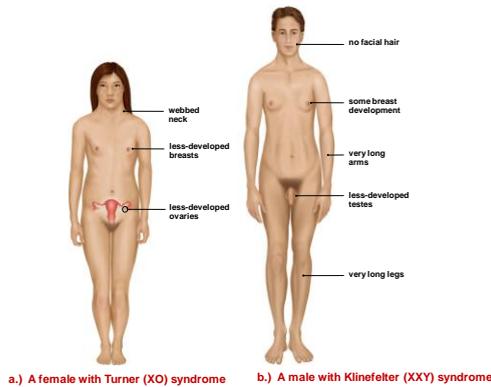
2. Sperm forming cells do not stop in meiosis I of metaphase

- Sperm cells don't get old
- Therefore, no nondisjunction

Abnormal sex chromosome number

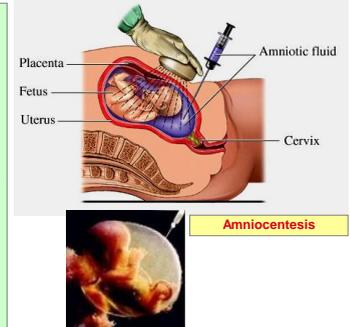
- Too few or too many X or Y chromosomes
- Newborns with abnormal sex chromosome numbers are more likely to survive than those with abnormal autosome numbers.
 - Extra X chromosomes become Barr bodies – inactivated
- Y determines maleness
 - SRY (sex-determining region Y) gene on Y chromosome
- Turner syndrome (45, XO)
 - Absence of second sex chromosome
 - Female
- Klinefelter syndrome (47, XXY)
 - Extra X inactivated as Barr body
 - Male

Figure 9.11 Abnormal sex chromosome number



Screening for Down Syndrome: Amniocentesis

1. Fetus located using ultrasound, needle inserted to remove amniotic fluid
 - Not performed until 16th week of pregnancy
2. Fluid contains fetal biochemicals and fetal cells from skin, respiratory tract, urinary tract
3. Culture cells for 1-2 weeks
4. Make Karyotype to detect abnormal chromosome numbers



Screening for Down Syndrome: Chorionic Villi Sampling (CVS)

1. Catheter inserted vaginally and chorionic tissue removed
2. Perform 9-11 weeks after conception
3. Make Karyotype
4. CVS –
 - Done earlier in pregnancy
 - Less chance of complications if end pregnancy
 - Slightly riskier for fetus
 - Greater chance of infection
5. Amniocentesis – have results...
 - Done later in pregnancy
 - Slightly safer than CVS



9.3 Mitosis Compared to Meiosis

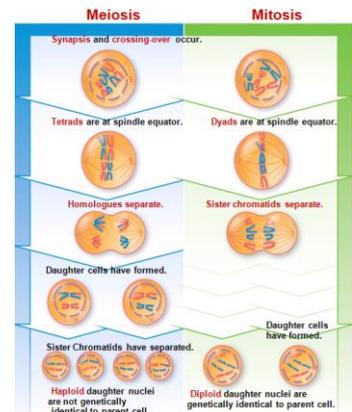
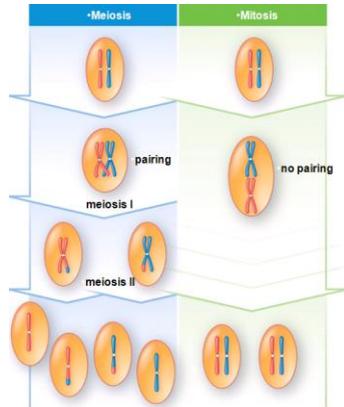


Figure 9.8

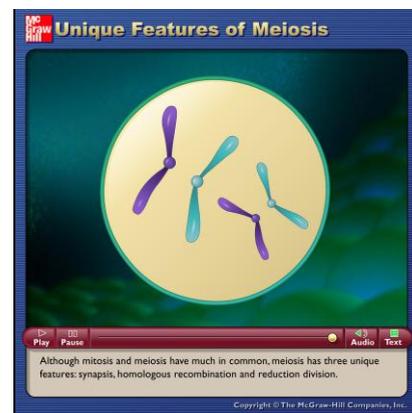
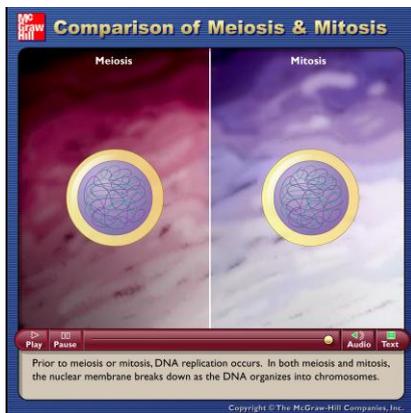
Meiosis II Compared to Mitosis

- Meiosis II is just like mitosis except that in meiosis II the cells have the haploid number of chromosomes.



9.3 Mitosis Compared to Meiosis

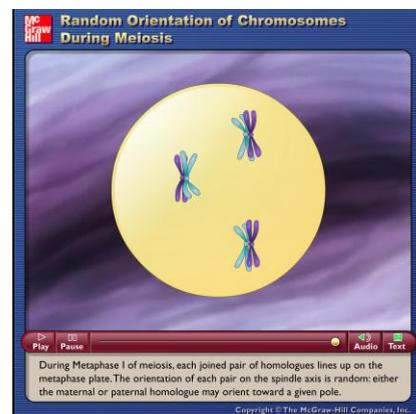
- | | |
|--|--|
| <ol style="list-style-type: none"> No cross-over Produces 2 genetically identical somatic cells Involves only 1 division Chromosomes (dyads) align single file in the middle of the cell during metaphase Sister chromatids separate during anaphase Daughter cells have the same number of chromosomes as the parent cell | <ol style="list-style-type: none"> Cross-over during prophase I Produces 4 genetically different gametes Involves 2 divisions Homologous pairs (tetrads) align during metaphase I Homologous pairs separate during anaphase I <ul style="list-style-type: none"> Sister chromatids separate during anaphase 2 Daughter cells have half the number of chromosomes as the parent cell |
|--|--|

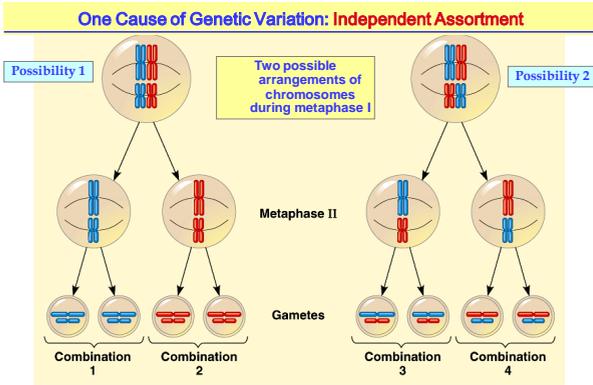


Why Meiosis Causes Genetic Variation

Independent Assortment

- Homologous pairs of chromosomes align independently of one another during metaphase I
 - Maternal and paternal chromosomes are shuffled during meiosis
 - 2^{23} or 8,388,608 different combinations for each parent
- Fertilization gives **70 trillion** possible genetic combinations





Cross-over—the 2nd Reason for Genetic Variation

- Homologous chromosomes exchange parts during Prophase I
- Results in thousands of genetically different gametes

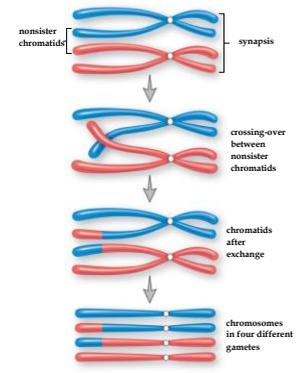
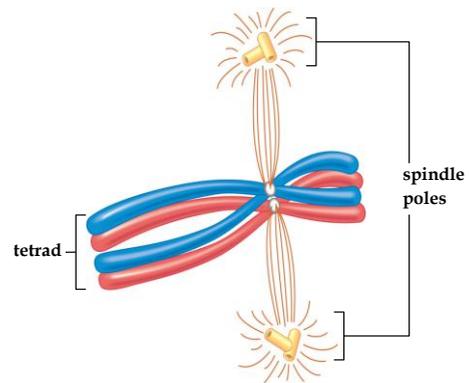


Figure 9.5
Crossing-over during prophase I

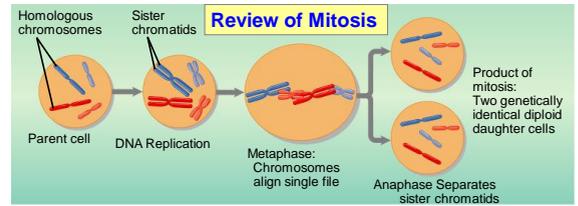
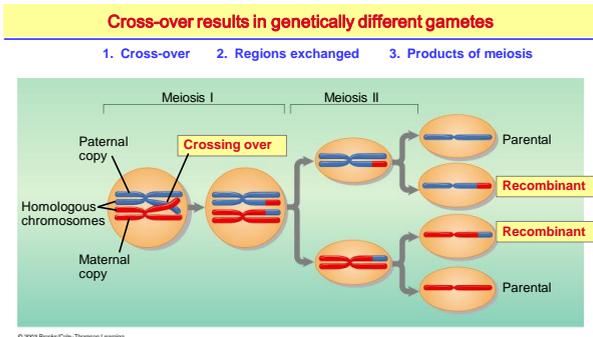
Figure 9.4 Synapsis



Meiosis with Crossing Over

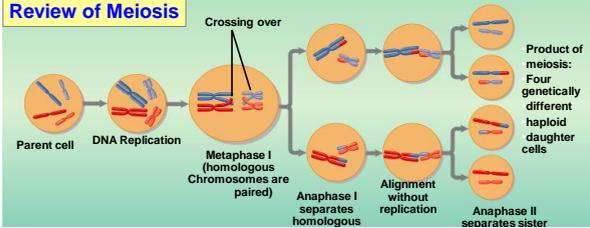
Crossing over during meiosis allows recombination of genes between homologous chromosomes. This alters the linkage between genes on the same chromosome.

Copyright © The McGraw-Hill Companies, Inc.



1. How do chromo's align at metaphase?
2. What separates at anaphase?
3. How do the daughter cells compare genetically?

Review of Meiosis



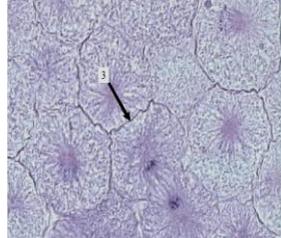
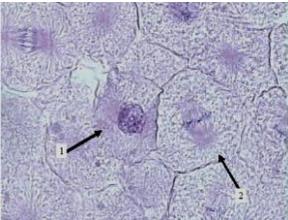
1. How do the chromo's align at metaphase I?
2. What separates at anaphase I?
3. How do the chromo's align at metaphase II?
4. What separates at anaphase II?

Mitosis and Meiosis Practice Problems

1. The phase of **mitosis** in which sister chromatids are separated is called
A. prophase. B. metaphase.
C. anaphase. D. telophase.
2. The phase of **mitosis** in which chromosomes condense is called
A. prophase. B. metaphase.
C. anaphase. D. telophase.
3. The phase of **meiosis** in which the nuclear membrane is dismantled is called
A. prophase I. B. anaphase I.
C. prophase II. D. metaphase II.
4. The phase of **meiosis** in which sister chromatids are separated is called
A. metaphase I. B. anaphase I.
C. anaphase II. D. metaphase I.
5. Most of the problems with chromosome numbers in cells are a result of
A. alcohol. B. U.V. light
C. non-disjunction. D. mitosis
6. List four differences between mitosis and meiosis.
7. Cite two ways that allow for genetic variation in an organism from meiosis.

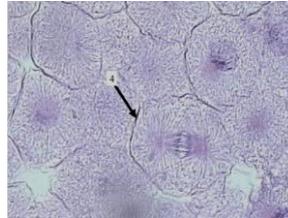
Mitosis Practice Problems

1. Identify the stage of mitosis for cell #1 below.
2. Identify the stage of mitosis for cell #2 below.
3. Identify the stage of mitosis for cell #3 below.
4. Identify the stage of mitosis for cell #4 below.



Mitosis Practice Problems

1. Identify the stage of mitosis for cell #4 below.



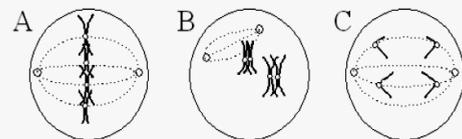
2. A diploid cell is one that
a. has two homologues of each chromosome.
b. is designated by the symbol $2n$.
c. has chromosomes found in pairs.
d. All of the above

Mitosis and Meiosis Practice Problems

1. During **anaphase of mitosis** in humans or other diploid organisms, how many chromatids does each chromosome have as they move toward the poles?
2. During **anaphase I of meiosis**, how many chromatids does each chromosome have as they move toward the poles?
3. During **anaphase II of meiosis**, how many chromatids does each chromosome have as they move toward the poles?
4. A student is simulating **meiosis I** with chromosomes that are red long and yellow long; red short and yellow short. Why would you not expect to find both red long and yellow long in one resulting daughter cell?
5. If there are 13 pairs of homologous chromosomes in a pre-sperm cell, how many chromosomes are there in a sperm? How many chromatids?

Mitosis and Meiosis Practice Problems

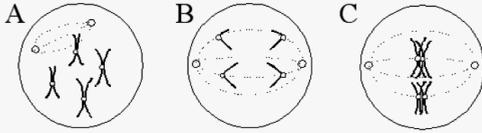
Below are three dividing cells. Assuming that all three cells come from the same species, use the first blank below each cell to tell whether this is **mitosis**, **meiosis I**, or **meiosis II** and the second blank to tell whether the cell is in **mitosis** or **meiosis** the cell is in.



division: _____
stage: _____

Mitosis and Meiosis Practice Problems

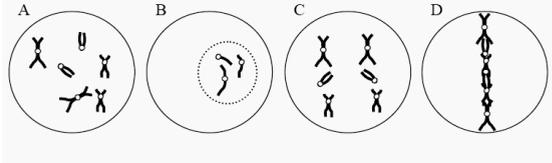
The figure below shows three cells in the process of division. If no additional information is given, can you tell what kind of division is occurring? Fill in the first blank under each cell with mitosis, meiosis I or meiosis II. If there is more than one possibility, list all of them. Then fill in the second blank to indicate the stage (phase) of mitosis or meiosis that this cell is in.



division: _____
 stage: _____

Mitosis and Meiosis Practice Problems

5. The cells below come from the same organism and are in the process of cell division:



- What is N for this organism?
- Circle the letter of each cell that could be either a germ cell or a gamete.
- In the blank below each cell, place one of the following letters: **I** for Interphase, **P** for Prophase, **M** for Metaphase, **A** for Anaphase or **T** for Telophase.