Chapter 19
Lecture Outline

See separate PowerPoint slides for all figures and tables pre-inserted into PowerPoint without notes.
Patterns of Chromosome Inheritance
Points to ponder

• What is the structure of chromosomes?
• What is the cell cycle and what occurs during each of its stages?
• Explain what mitosis is used for, what cells undergo mitosis, and the four stages of mitosis.
• Explain the two divisions of meiosis.
• What is meiosis used for and what cells undergo meiosis?
• Compare and contrast mitosis and meiosis.
• Compare and contrast spermatogenesis and oogenesis.
• What are trisomy and monosomy?
• What most often causes changes in chromosome number?
• What are the syndromes associated with changes in sex chromosomes?
• Explain the four changes in chromosome structure.
Chromosomes: A review

• Humans have 46 chromosomes that are in 23 pairs within a cell’s nucleus.
  – Pairs of chromosomes are called homologous chromosomes.
  – **Autosomes** are the 22 pairs of chromosomes that control traits unrelated to gender.
  – **Sex chromosomes** are the 1 pair that contains the genes that do control gender.
Chromosomes: A review

- Cells (body cells) that have 46 (2n) paired chromosomes are called *diploid*.

- Cells (sex cells) that have only 23 (n) unpaired chromosomes are called *haploid*.
What is a karyotype?

The 46 chromosomes of a male

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Figure 19.1 A karyotype of human chromosomes.
The cell cycle

• Two parts

1. Interphase
   • $G_1$ stage – cell doubles its organelles; cell grows in size
   • S stage – DNA replication occurs
   • $G_2$ stage – proteins needed for division are synthesized

2. Cell division (mitosis and cytokinesis)
   • Mitosis – nuclear division
   • Cytokinesis – cytoplasmic division
The cell cycle

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Figure 19.2 Stages of the cell cycle.
Control of the cell cycle

- **Checkpoints** can delay the cell cycle until certain conditions are met.

- There are three major checkpoints.
  - $G_1$, if passed, the cell is committed to divide.
  - $G_2$ cycle pauses to verify DNA replication.
  - $M$ cycle pauses to verify spindle assembly and chromosome attachment.
19.2 The Cell Cycle

Control of the cell cycle

**G₁ checkpoint**
Cell cycle main checkpoint. If DNA is damaged, apoptosis will occur. Otherwise, the cell is committed to divide when growth signals are present and nutrients are available.

**G₂ checkpoint**
Mitosis checkpoint. Mitosis will occur if DNA has replicated properly. Apoptosis will occur if the DNA is damaged and cannot be repaired.

**M checkpoint**
Spindle assembly checkpoint. Mitosis will not continue if chromosomes are not properly aligned.

*Figure 19.3 Control of the cell cycle.*
Chromosome structure in mitosis

• Chromosomes contain both DNA and proteins (collectively called chromatin).

• Chromosomes that are dividing are made up of two identical parts called sister chromatids.

• The sister chromatids are held together at a region called the centromere.
Chromosome structure in mitosis

- Chromosome
- Replication
- Division
- Sister chromatids
- Centromere
- Duplicated chromosome
The spindle in mitosis

- **Centrosome** – the microtubule organizing center of the cell

- **Aster** – an array of microtubules at one of the poles (ends of the cell)

- **Centrioles** – short cylinders of microtubules that assist in the formation of spindle fibers
The spindle in mitosis

19.3 Mitosis

Figure 19.7 The mitotic spindle.
Overview of mitosis

- A diploid cell makes and divides an exact copy of its nucleus.
- It is used in cell growth and cell repair.
- Mitosis occurs in body cells.
- There are four phases.
  1. Prophase
  2. Metaphase
  3. Anaphase
  4. Telophase
19.3 Mitosis

Figure 19.6 An overview of mitosis.
19.3 Mitosis

1. Mitosis: Prophase

- Chromosomes condense and become visible.
- The nuclear envelope fragments.
- The nucleolus disappears.
- Centrosomes move to opposite poles.
- Spindle fibers appear and attach to the centromeres.
1. Mitosis: Prophase

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Figure 19.8 The phases of mitosis.

(early prophase, prophase): © Ed Reschke
2. Mitosis: Metaphase

- Chromosomes line up at the middle of the cell (equator).
- Spindle becomes fully-formed.

Figure 19.8 The phases of mitosis.
3. Mitosis: Anaphase

- Sister chromatids separate at the centromeres and move towards the poles.

Figure 19.8 The phases of mitosis.
4. Mitosis: Telophase and cytokinesis

- Chromosomes arrive at the poles.
- Chromosomes become indistinct chromatin again.
- Nucleoli reappear.
- Spindle disappears.
- Nuclear envelope reassembles.
- Two daughter cells are formed by a ring of actin filaments (cleavage furrow).

*Figure 19.8 The phases of mitosis.*
Overview of meiosis

- Two nuclear divisions occur to make four haploid cells.
- It is used to make gametes (egg and sperm).
- **Meiosis** occurs in sex cells.
- It has eight phases (4 in each meiosis, I & II).

**Figure 19.9** The results of meiosis.
Meiosis I

- **Prophase I**
  - Homologous chromosomes pair (synapsis); crossing-over occurs, in which there is exchange of genetic information.

- **Metaphase I**
  - Homologous pairs line up at the equator.

- **Anaphase I**
  - Homologous chromosomes separate and move toward opposite poles.

- **Telophase I**
  - Two daughter cells result, each with 23 duplicated chromosomes.
19.4 Meiosis

Meiosis I

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Prophase I
Chromosomes have duplicated. Homologous chromosomes pair during synapsis and crossing-over occurs.

Figure 19.10 The phases of meiosis.
Meiosis I

Prophase I
Chromosomes have duplicated. Homologous chromosomes pair during synapsis and crossing-over occurs.

Metaphase I
Homologous pairs align independently at the equator.

Figure 19.10 The phases of meiosis.
19.4 Meiosis

Meiosis I

Prophase I
Chromosomes have duplicated. Homologous chromosomes pair during synapsis and crossing-over occurs.

Diploid

Metaphase I
Homologous pairs align independently at the equator.

Anaphase I
Homologous chromosomes separate and move toward the poles.

Figure 19.10 The phases of meiosis.
19.4 Meiosis

Meiosis I

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Figure 19.10 The phases of meiosis.

**Meiosis I**

**Prophase I**
Homologous chromosomes pair during synapsis.

**Metaphase I**
Homologous chromosome pairs align at the metaphase plate.

**Anaphase I**
Homologous chromosomes separate, pulled to opposite poles by centromeric spindle fibers.

**Telophase I**
Daughter cells have one chromosome from each homologous pair.
Figure 19.10 The phases of meiosis.
What is crossing-over?

- **Crossing-over** is the exchange of genetic information between nonhomologous sister chromatids during synapsis.

- This occurs during prophase I of meiosis and increases genetic variation.
What is crossing-over?

Figure 19.11  Synapsis and crossing-over increase variability.
What is independent alignment?

- In metaphase I, homologous pairs align independently at the equator.

- Maternal or paternal member may be oriented toward either pole.

- It gives rise to various possible combinations of maternal and paternal chromosomes in gametes descended from this cell.
Independent alignment at metaphase I increases variability.
Meiosis II

- **Prophase II**
  - Chromosomes condense again.

- **Metaphase II**
  - Chromosomes align at the equator.

- **Anaphase II**
  - Sister chromatids separate to opposite poles.

- **Telophase II**
  - Four daughter cells result, each with 23 unduplicated chromosomes.
Meiosis II

Figure 19.10 The phases of meiosis.
19.4 Meiosis

Meiosis II

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**Cells from Meiosis I**

![Cells from Meiosis I](image)

**Prophase II**
Cells have one chromosome from each homologous pair.

**Metaphase II**
Chromosomes align at the metaphase plate.

**Figure 19.10** The phases of meiosis.
Figure 19.10 The phases of meiosis.

19.4 Meiosis

Meiosis II

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MEIOSIS II
Prophase II
Cells have one chromosome from each homologous pair.

Metaphase II
Chromosomes align at the metaphase plate.

Anaphase II
Sister chromatids separate and move toward the poles.
19.4 Meiosis

Meiosis II

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Figure 19.10 The phases of meiosis.
Meiosis II

Cells from Meiosis I

Prophase II
Cells have one chromosome from each homologous pair.

Metaphase II
Chromosomes align at the metaphase plate.

Anaphase II
Sister chromatids separate and move toward the poles.

Telophase II
Spindle disappears, nuclei form, and cytokinesis takes place.

Daughter Cells
Meiosis results in four haploid daughter cells.

Figure 19.10  The phases of meiosis.
The production of sperm and eggs

• **Spermatogenesis**
  – It is the process of making sperm in males.
  – It is a continual process after puberty.
  – About 400 million sperm are produced per day.
19.4 Meiosis

The production of sperm and eggs

- **Oogenesis**
  - The process of making eggs in females.
  - During meiosis, one egg and three polar bodies are formed.
  - Polar bodies act to hold discarded chromosomes and thus disintegrate.
  - Normally, one egg per month is produced, and 500 are produced during the entire reproductive cycle.
19.4 Meiosis

The production of sperm and eggs

Figure 19.13 A comparison of spermatogenesis and oogenesis in mammals.
19.5 Comparison of Meiosis and Mitosis

**Mitosis** vs. **Meiosis**

- Growth and repair of cells
- Occurs in body cells
- 1 division
- Results in 2 diploid, genetically identical cells

- Formation of gametes
- Occurs in sex cells
- 2 divisions
- Results in 4 haploid, genetically different cells
18.5 Comparison of Meiosis and Mitosis

Comparing meiosis and mitosis

Figure 19.14 A comparison of meiosis and mitosis.
Comparing meiosis and mitosis

**Figure 19.14** A comparison of meiosis and mitosis.

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**Comparing meiosis and mitosis**

- **Meiosis**
  - Telophase I: Daughter cells form.
  - Meiosis II: Sister chromatids separate.
  - Daughter nuclei are not genetically identical to parental cell.

- **Mitosis**
  - Telophase: Daughter cells form.
  - Daughter nuclei are genetically identical to parental cell.
Changes in chromosome number

- **Nondisjunction** occurs when both members of a homologous pair go into the same daughter cell during meiosis I, or when sister chromatids fail to separate in meiosis II.

- **Results of nondisjunction**
  - **Monosomy**: cell has only 1 copy of a chromosome
    e.g., Turner syndrome (only 1 X chromosome)
  - **Trisomy**: cell has 3 copies of a chromosome
    e.g., Down syndrome (3 copies of chromosome 21)
Figure 19.15 The consequences of nondisjunction of chromosomes during oogenesis.
Barr bodies and X inactivation

• Females have 2 copies of X genes; males have 1.

• In female embryos, 1 X chromosome becomes inactivated (Barr body) in each cell.
Changes in chromosome number

Figure 19.16 Down syndrome.
Changes in sex chromosome number

- **Turner syndrome** (XO) – short stature, broad-shouldered, with folds of skin on the neck, underdeveloped sex organs and breasts

- **Klinefelter syndrome** (XXY) – underdeveloped sex organs, breast development, large hands, and long arms and legs
Changes in sex chromosome number

- **Poly-X females** (XXX, XXXX)
  - XXX tend to be tall and thin but do not usually have mental retardation
  - XXXX have severe mental retardation

- **Jacobs syndrome** (XYY) – tall, persistent acne, speech and reading problems
Changes in chromosome structure

- **Deletion** – loss of a piece of the chromosome (e.g., Williams syndrome)

- **Translocation** – movement of chromosome segments from one chromosome to another nonhomologous chromosome (Alagille syndrome)

- **Duplication** – presence of a chromosome segment more than once in the same chromosome

- **Inversion** – a segment of a chromosome is inverted 180 degrees
19.6 Chromosome Inheritance

Changes in chromosome structure

![Diagram showing various types of chromosomal mutations: a. Deletion, b. Duplication, c. Inversion, d. Translocation.]

**Figure 19.17** The various types of chromosomal mutations.
Chromosomal inversion

Figure 19.18 A chromosomal inversion.
19.6 Chromosome Inheritance

Changes in chromosome structure

Figure 19.19 A chromosomal deletion.

Figure 19.20 A chromosomal translocation.