Chapter 17
Chromosomal Inheritance

Outline
- Human Life Cycle
  - Mitosis
    - Stages of Mitosis
  - Meiosis
    - Crossing-Over
    - Stages of Meiosis
  - Chromosomal Inheritance
  - Autosomal Syndromes
  - Sex Chromosomal Syndromes

Human Life Cycle
- Mitosis ensures every cell has a complete number of chromosomes.
- Meiosis reduces the chromosome number by half in gametes.
  - Sperm and egg (gametes) are haploid (n).
  - Somatic cells are diploid (2n).

Mitosis
- Mitosis occurs in humans when tissues grow or when repair occurs, and produces daughter cells with the same number of chromosomes as the parental cells.
  - Sister chromosomes separate, and one of each kind of chromosome goes into each daughter cell.

Mitosis Overview

Cell Cycle
- Cell cycle consists of:
  - Interphase.
  - Mitosis.
    - Cytokinesis.
  - Interphase is the interval of time between cell divisions, and is the phase the cell is in the longest.

Stages of Mitosis
- Prophase.
  - Centrioles outside nucleus move away from each other.
  - Spindle fibers appear.
  - Nuclear envelope fragments.
- Metaphase.
  - Spindle fully-formed.
- Chromosomes align at the equator.

9 Stages of Mitosis
- Anaphase.
  - Sister chromosomes separate and daughter chromosomes move to the poles.
  - Spindle fibers shorten and pull chromosomes towards the poles.
- Telophase.
  - Chromosomes arrive at the poles.
  - Chromosomes become indistinct chromatin.
  - Two daughter cells.

10 Meiosis
- Meiosis requires two nuclear divisions and results in four daughter cells, each with half the number of parental chromosomes.
  - Humans have 23 pairs of homologous chromosomes.
  - During meiosis I synapsis occurs allowing crossing-over.
    - Exchange of genetic material between chromatids.

12 Crossing-Over

13 Meiosis
- At the beginning of Meiosis II, chromosomes are dyads because each is composed of two sister chromatids.
- During Meiosis II, sister chromatids separate in each of the cells from Meiosis I.
  - Each of the resulting four daughter cells has the haploid number of chromosomes.

14 Meiosis Overview

15 The Importance of Meiosis
- Three ways individuals are assured a different genetic combination than either parent.
  - Crossing-over recombines genes on sister chromosomes of homologous pairs.
  - Following meiosis, gametes have all possible chromosome combinations.
  - At fertilization, sperm and egg carry varied chromosome combinations.

16 Spermatogenesis and Oogenesis
- Spermatogenesis occurs in the testes of males and produces haploid sperm.
  - Once started, continues to completion.
- Oogenesis occurs in the ovaries of females, and produces haploid eggs.
  - Does not necessarily go to completion.
Stages of Meiosis
- First Division.
  - Prophase I - Spindle appears, and nuclear envelope fragments.
  - Metaphase I - Tetradis line up at equator.
  - Anaphase I - Homologous chromosomes of each pair separate and 
    move to opposite poles of the spindle.
  - Telophase I - Spindle disappears and nuclear envelope reforms.
  - Cytokinesis - Plasma membrane furrows.

Stages of Meiosis
- Second Division.
  - Prophase II - Spindle appears and nuclear envelope disassembles.
  - Metaphase II - Dyads line up at equator.
  - Anaphase II - Sister chromatids separate and move towards 
    poles.
  - Telophase II - Spindle disappears and nuclear envelope reforms.
  - Cytokinesis - Plasma membrane furrows.
  * Four haploid daughter cells produced.

Chromosomal Inheritance
- Humans have 22 pairs of autosomes, and one pair of sex chromosomes.
  - Abnormal chromosome number or structure often leads to a syndrome.
  - Amniocentesis and chorionic villi sampling can be used to obtain a genetic 
    sample to produce a karyotype.
  - Visual display of chromosomes arranged by size, shape, and banding 
    pattern.

Human Karyotype Preparation

Autosomal Syndromes
- Nondisjunction occurs:
  - During Meiosis I when both members of a homologous pair go to the 
    same daughter cell.
  - During Meiosis II when sister chromosomes fail to separate and both 
    daughter chromosomes go to the same gamete.

Down Syndrome.
  - Trisomy 21.
  - Incidence increases with mother's age.
- Cri du Chat Syndrome.
  - Chromosomal deletion.

Sex Chromosomal Syndromes
- Fragile X Syndrome.
- Abnormal Sex Chromosome Number.
  - Turner Syndrome.
    - XO.
  - Klinefelter Syndrome.
    - Two or more X chromosomes with a Y.
  - Poly-X Females.
    - More than two X chromosomes.
  - Jacobs Syndrome.
    - XYY.

25 Turner and Klinefelter Syndromes

26 Review
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  - Autosomal Syndromes
  - Sex Chromosomal Syndromes