Section A: An Introduction to Heredity

1. Offspring acquire genes from parents by inheriting chromosomes
2. Like begets like, more or less: a comparison of asexual and sexual reproduction
Living organisms are distinguished by their ability to reproduce their own kind.

Offspring resemble their parents more than they do less closely related individuals of the same species.

The transmission of traits from one generation to the next is called heredity or inheritance.

However, offspring differ somewhat from parents and siblings, demonstrating variation.

Genetics is the study of heredity and variation.
1. Offspring acquire genes from parents by inheriting chromosomes

- Parents endow their offspring with coded information in the form of genes.
  - Your genome is derived from the thousands of genes that you inherited from your mother and your father.

- Genes program specific traits that emerge as we develop from fertilized eggs into adults.
  - Your genome may include a gene for freckles, which you inherited from your mother.
• Genes are segments of DNA.

• Genetic information is transmitted as specific sequences of the four deoxyribonucleotides in DNA.
  
  • This is analogous to the symbolic information of letters in which words and sentences are translated into mental images.
  
  • Cells translate genetic “sentences” into freckles and other features with no resemblance to genes.

• Most genes program cells to synthesize specific enzymes and other proteins that produce an organism’s inherited traits.
The transmission of hereditary traits has its molecular basis in the precise replication of DNA.

- This produces copies of genes that can be passed from parents to offspring.

In plants and animals, sperm and ova (unfertilized eggs) transmit genes from one generation to the next.

After fertilization (fusion) of a sperm cell with an ovum, genes from both parents are present in the nucleus of the fertilized egg.
Almost all of the DNA in a eukaryotic cell is subdivided into chromosomes in the nucleus.

- Tiny amounts of DNA are found in mitochondria and chloroplasts.

Every living species has a characteristic number of chromosomes.

- Humans have 46 in almost all of their cells.

Each chromosome consists of a single DNA molecule in association with various proteins.

Each chromosome has hundreds or thousands of genes, each at a specific location, its locus.
• In asexual reproduction, a single individual passes along copies of all its genes to its offspring.
  
  • Single-celled eukaryotes reproduce asexually by mitotic cell division to produce two identical daughter cells.

• Even some multicellular eukaryotes, like hydra, can reproduce by budding cells produced by mitosis.

Fig. 13.1
• Sexual reproduction results in greater variation among offspring than does asexual reproduction.

• Two parents give rise to offspring that have unique combinations of genes inherited from the parents.

• Offspring of sexual reproduction vary genetically from their siblings and from both parents.
Section B: The Role of Meiosis in Sexual Life Cycles

1. Fertilization and meiosis alternate in sexual life cycles
2. Meiosis reduces chromosome number from diploid to haploid: *a closer look*
Introduction

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

• It starts at the conception of an organism and continues until it produces its own offspring.
1. Fertilization and meiosis alternate in sexual life cycles

- In humans, each somatic cell (all cells other than sperm or ovum) has 46 chromosomes.
  - Each chromosome can be distinguished by its size, position of the centromere, and by pattern of staining with certain dyes.
- A karyotype display of the 46 chromosomes shows 23 pairs of chromosomes, each pair with the same length, centromere position, and staining pattern.
- These homologous chromosome pairs carry genes that control the same inherited characters.
• Karyotypes, ordered displays of an individual’s chromosomes, are often prepared with lymphocytes.
• An exception to the rule of homologous chromosomes is found in the sex chromosomes, the X and the Y.

• The pattern of inheritance of these chromosomes determines an individual’s sex.
  • Human females have a homologous pair of X chromosomes (XX).
  • Human males have an X and a Y chromosome (XY).

• Because only small parts of these have the same genes, most of their genes have no counterpart on the other chromosome.

• The other 22 pairs are called autosomes.
The occurrence of homologous pairs of chromosomes is a consequence of sexual reproduction.

We inherit one chromosome of each homologous pair from each parent.

- The 46 chromosomes in a somatic cell can be viewed as two sets of 23, a maternal set and a paternal set.

Sperm cells or ova (gametes) have only one set of chromosomes - 22 autosomes and an X or a Y.

A cell with a single chromosome set is haploid.

- For humans, the haploid number of chromosomes is 23 (n = 23).
• By means of sexual intercourse, a haploid sperm reaches and fuses with a haploid ovum.

• These cells fuse (syngamy) resulting in fertilization.

• The fertilized egg (zygote) now has two haploid sets of chromosomes bearing genes from the maternal and paternal family lines.

• The zygote and all cells with two sets of chromosomes are diploid cells.
  • For humans, the diploid number of chromosomes is 46 (2n = 46).
• As an organism develops from a zygote to a sexually mature adult, the zygote’s genes are passed on to all somatic cells by mitosis.

• Gametes, which develop in the gonads, are not produced by mitosis.
  • If gametes were produced by mitosis, the fusion of gametes would produce offspring with four sets of chromosomes after one generation, eight after a second and so on.

• Instead, gametes undergo the process of **meiosis** in which the chromosome number is halved.
  • Human sperm or ova have a haploid set of 23 different chromosomes, one from each homologous pair.
• Fertilization restores the diploid condition by combining two haploid sets of chromosomes.

• Fertilization and meiosis alternate in sexual life cycles.
The timing of meiosis and fertilization does vary among species.

The life cycle of humans and other animals is typical of one major type.

- Gametes, produced by meiosis, are the only haploid cells.
- Gametes undergo no divisions themselves, but fuse to form a diploid zygote that divides by mitosis to produce a multicellular organism.
Most fungi and some protists have a second type of life cycle.

- The zygote is the only diploid phase.
- After fusion of two gametes to form a zygote, the zygote undergoes meiosis to produce haploid cells.
- These haploid cells undergo mitosis to develop into a haploid multicellular adult organism.
- Some haploid cells develop into gametes by mitosis.

Fig. 13.5b
Plants and some algae have a third type of life cycle, **alternation of generations**.

- This life cycle includes both haploid (gametophyte) and diploid (sporophyte) multicellular stages.
- Meiosis by the sporophyte produces haploid **spores** that develop by mitosis into the gametophyte.
- Gametes produced via mitosis by the gametophyte fuse to form the zygote which produces the sporophyte by mitosis.
3. Meiosis reduces chromosome number from diploid to haploid: *a closer look*

- Many steps of meiosis resemble steps in mitosis.
- Both are preceded by the replication of chromosomes.
- However, in meiosis, there are two consecutive cell divisions, *meiosis I* and *meiosis II*, that result in four daughter cells.
- Each final daughter cell has only half as many chromosomes as the parent cell.
• Meiosis reduces chromosome number by copying the chromosomes once, but dividing twice.

• The first division, meiosis I, separates homologous chromosomes.

• The second, meiosis II, separates sister chromatids.
• Division in meiosis I occurs in four phases: prophase, metaphase, anaphase, and telophase.

• During the preceding interphase the chromosomes are replicated to form sister chromatids.
  
  • These are genetically identical and joined at the centromere.

• Also, the single centrosome is replicated.

Fig. 13.7
• In prophase I, the chromosomes condense and homologous chromosomes pair up to form tetrads.
  • In a process called synapsis, special proteins attach homologous chromosomes tightly together.
  • At several sites the chromatids of homologous chromosomes are crossed (chiasmata) and segments of the chromosomes are traded.
  • A spindle forms from each centrosome and spindle fibers attached to kinetochores on the chromosomes begin to move the tetrads around.
At metaphase I, the tetrads are all arranged at the metaphase plate.

Microtubules from one pole are attached to the kinetochore of one chromosome of each tetrad, while those from the other pole are attached to the other.

In anaphase I, the homologous chromosomes separate and are pulled toward opposite poles.
• In telophase I, movement of homologous chromosomes continues until there is a haploid set at each pole.
  • Each chromosome consists of linked sister chromatids.

• Cytokinesis by the same mechanisms as mitosis usually occurs simultaneously.

• In some species, nuclei may reform, but there is no further replication of chromosomes.
Meiosis II is very similar to mitosis.

During prophase II a spindle apparatus forms, attaches to kinetochores of each sister chromatid, and moves them around.

Spindle fibers from one pole attach to the kinetochore of one sister chromatid and those of the other pole to the other sister chromatid.
• At metaphase II, the sister chromatids are arranged at the metaphase plate.
  • The kinetochores of sister chromatids face opposite poles.
• At anaphase II, the centomeres of sister chromatids separate and the now separate sisters travel toward opposite poles.
• In telophase II, separated sister chromatids arrive at opposite poles.
  • Nuclei form around the chromatids.

• Cytokinesis separates the cytoplasm.

• At the end of meiosis, there are four haploid daughter cells.

Fig. 13.7
Mitosis and meiosis have several key differences.

- The chromosome number is reduced by half in meiosis, but not in mitosis.
- Mitosis produces daughter cells that are genetically identical to the parent and to each other.
- Meiosis produces cells that differ from the parent and each other.
Three events, unique to meiosis, occur during the first division cycle.

1. During prophase I, homologous chromosomes pair up in a process called synapsis.

   - A protein zipper, the synaptonemal complex, holds homologous chromosomes together tightly.
   - Later in prophase I, the joined homologous chromosomes are visible as a tetrad.
   - At X-shaped regions called chiasmata, sections of nonsister chromatids are exchanged.
   - Chiasmata is the physical manifestation of crossing over, a form of genetic rearrangement.
2. At metaphase I homologous pairs of chromosomes, not individual chromosomes are aligned along the metaphase plate.
   • In humans, you would see 23 tetrads.

3. At anaphase I, it is homologous chromosomes, not sister chromatids, that separate and are carried to opposite poles of the cell.
   • Sister chromatids remain attached at the centromere until anaphase II.
   • The processes during the second meiotic division are virtually identical to those of mitosis.
Mitosis produces two identical daughter cells, but meiosis produces 4 very different cells.
<table>
<thead>
<tr>
<th>Event</th>
<th>Mitosis</th>
<th>Meiosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>DNA replication</td>
<td>Occurs during interphase before nuclear division begins</td>
<td>Occurs once, during the interphase before meiosis I begins</td>
</tr>
<tr>
<td>Number of divisions</td>
<td>One, including prophase, metaphase, anaphase, and telophase</td>
<td>Two, each including prophase, metaphase, anaphase, and telophase</td>
</tr>
<tr>
<td>Synapsis of homologous</td>
<td>Does not occur</td>
<td>Synapsis is unique to meiosis: During prophase I, the homologous</td>
</tr>
<tr>
<td>chromosomes</td>
<td></td>
<td>chromosomes join along their length, forming tetrads (groups of four</td>
</tr>
<tr>
<td></td>
<td></td>
<td>chromatids); synopsis is associated with crossing over between</td>
</tr>
<tr>
<td></td>
<td></td>
<td>nonsister chromatids</td>
</tr>
<tr>
<td>Number of daughter cells</td>
<td>Two, each diploid (2n) and genetically identical to the parent cell</td>
<td>Four, each haploid (n), containing half as many chromosomes as the</td>
</tr>
<tr>
<td>and genetic composition</td>
<td></td>
<td>parent cell; genetically nonidentical to the parent cell and to each</td>
</tr>
<tr>
<td>Role in the animal body</td>
<td>Enables multicellular adult to arise from zygote; produces cells for</td>
<td>Produces gametes; reduces chromosome number by half and introduces</td>
</tr>
<tr>
<td></td>
<td>growth and tissue repair</td>
<td>genetic variability among the gametes</td>
</tr>
</tbody>
</table>
Section C: Origins of Genetic Variation

1. Sexual life cycles produce genetic variation among offspring
2. Evolutionary adaptation depends on a population’s genetic variation
1. Sexual life cycles produce genetic variation among offspring

- The behavior of chromosomes during meiosis and fertilization is responsible for most of the variation that arises each generation during sexual reproduction.

- Three mechanisms contribute to genetic variation:
  - independent assortment
  - crossing over
  - random fertilization
• Independent assortment of chromosomes contributes to genetic variability due to the random orientation of tetrads at the metaphase plate.

• There is a fifty-fifty chance that a particular daughter cell of meiosis I will get the maternal chromosome of a certain homologous pair and a fifty-fifty chance that it will receive the paternal chromosome.
• Each homologous pair of chromosomes is positioned independently of the other pairs at metaphase I.

• Therefore, the first meiotic division results in independent assortment of maternal and paternal chromosomes into daughter cells.

• The number of combinations possible when chromosomes assort independently into gametes is $2^n$, where $n$ is the haploid number of the organism.

  • If $n = 3$, there are eight possible combinations.

  • For humans with $n = 23$, there are $2^{23}$ or about 8 million possible combinations of chromosomes.
Independent assortment alone would find each individual chromosome in a gamete that would be exclusively maternal or paternal in origin.

However, crossing over produces recombinant chromosomes, which combine genes 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.
  • For humans, this occurs two to three times per chromosome pair.

• One sister chromatid may undergo different patterns of crossing over than its match.

• Independent assortment of these nonidentical sister chromatids during meiosis II increases still more the number of genetic types of gametes that can result from meiosis.
• The random nature of fertilization adds to the genetic variation arising from meiosis.

• Any sperm can fuse with any egg.
  • A zygote produced by a mating of a woman and man has a unique genetic identity.
  • An ovum is one of approximately 8 million possible chromosome combinations (actually $2^{23}$).
  • The successful sperm represents one of 8 million different possibilities (actually $2^{23}$).
  • The resulting zygote is composed of 1 in 70 trillion ($2^{23} \times 2^{23}$) possible combinations of chromosomes.
  • Crossing over adds even more variation to this.
The three sources of genetic variability in a sexually reproducing organism are:

- Independent assortment of homologous chromosomes during meiosis I and of nonidentical sister chromatids during meiosis II.
- Crossing over between homologous chromosomes during prophase I.
- Random fertilization of an ovum by a sperm.

All three mechanisms reshuffle the various genes carried by individual members of a population.

Mutations, still to be discussed, are what ultimately create a population’s diversity of genes.
2. Evolutionary adaptation depends on a population’s genetic variation

- Darwin recognized the importance of genetic variation in evolution via natural selection.
- A population evolves through the differential reproductive success of its variant members.
- Those individuals best suited to the local environment leave the most offspring, transmitting their genes in the process.
- This natural selection results in adaptation, the accumulation of favorable genetic variations.
As the environment changes or a population moves to a new environment, new genetic combinations that work best in the new conditions will produce more offspring and these genes will increase.

- The formerly favored genes will decrease.

Sex and mutations are two sources of the continual generation of new genetic variability.

Gregor Mendel, a contemporary of Darwin, published a theory of inheritance that helps explain genetic variation.

- However, this work was largely unknown for over 40 years until 1900.