

Overview: Variations on a Theme

- Living organisms are distinguished by their ability to reproduce their own kind
- **Genetics** is the scientific study of heredity and variation
- **Heredity** is the transmission of traits from one generation to the next
- **Variation** is demonstrated by the differences in appearance that offspring show from parents and siblings

Concept 13.1: Offspring acquire genes from parents by inheriting chromosomes

- In a literal sense, children do not inherit particular physical traits from their parents
- It is genes that are actually inherited

Inheritance of Genes

- **Genes** are the units of heredity, and are made up of segments of DNA
- Genes are passed to the next generation via reproductive cells called **gametes** (sperm and eggs)
- Each gene has a specific location called a **locus** on a certain chromosome
- Most DNA is packaged into chromosomes

Comparison of Asexual and Sexual Reproduction

- In **asexual reproduction**, a single individual passes genes to its offspring without the fusion of gametes
- A **clone** is a group of genetically identical individuals from the same parent
- In **sexual reproduction**, two parents give rise to offspring that have unique combinations of genes inherited from the two parents

Concept 13.2: Fertilization and meiosis alternate in sexual life cycles

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

Sets of Chromosomes in Human Cells

- Human **somatic cells** (any cell other than a gamete) have 23 pairs of chromosomes
- A **karyotype** is an ordered display of the pairs of chromosomes from a cell
- The two chromosomes in each pair are called **homologous chromosomes**, or homologs
- Chromosomes in a homologous pair are the same length and shape and carry genes controlling the same inherited characters
- The **sex chromosomes**, which determine the sex of the individual, are called X & Y
- Human females have a homologous pair of X chromosomes (XX)
- Human males have one X and one Y chromosome
- The remaining 22 pairs of chromosomes are called **autosomes**
- Each pair of homologous chromosomes includes one chromosome from each parent
- The 46 chromosomes in a human somatic cell are two sets of 23: one from the mother and one from the father
- A **diploid cell** ($2n$) has two sets of chromosomes
- For humans, the diploid number is 46 ($2n = 46$)
- In a cell in which DNA synthesis has occurred, each chromosome is replicated

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- Each replicated chromosome consists of two identical sister chromatids
- A gamete (sperm or egg) contains a single set of chromosomes, and is **haploid** (n)
- For humans, the haploid number is 23 ($n = 23$)
- Each set of 23 consists of 22 autosomes and a single sex chromosome
- In an unfertilized egg (ovum), the sex chromosome is X
- In a sperm cell, the sex chromosome may be either X or Y

- **Fertilization** is the union of gametes (the sperm and the egg)
- The fertilized egg is called a **zygote** and has one set of chromosomes from each parent
- The zygote produces somatic cells by mitosis and develops into an adult
- At sexual maturity, the ovaries and testes produce haploid gametes
- Gametes are the only types of human cells produced by **meiosis**, rather than mitosis
- Meiosis results in one set of chromosomes in each gamete
- Fertilization and meiosis alternate in sexual life cycles to maintain chromosome number

The Variety of Sexual Life Cycles

- The alternation of meiosis and fertilization is common to all organisms that reproduce sexually
- The three main types of sexual life cycles differ in the timing of meiosis and fertilization
- Gametes are the only haploid cells in animals
- They are produced by meiosis and undergo no further cell division before fertilization
- Gametes fuse to form a diploid zygote that divides by mitosis to develop into a multicellular organism
- Plants and some algae exhibit an **alternation of generations**
- This life cycle includes both a diploid and haploid multicellular stage
- The diploid organism, called the sporophyte, makes haploid spores by meiosis
- Each spore grows by mitosis into a haploid organism called a gametophyte
- A gametophyte makes haploid gametes by mitosis
- Fertilization of gametes results in a diploid sporophyte
- In most fungi and some protists, the only diploid stage is the single-celled zygote; there is no multicellular diploid stage
- The zygote produces haploid cells by meiosis
- Each haploid cell grows by mitosis into a haploid multicellular organism
- The haploid adult produces gametes by mitosis
- Depending on the type of life cycle, either haploid or diploid cells can divide by mitosis
- However, only diploid cells can undergo meiosis
- In all three life cycles, the halving and doubling of chromosomes contributes to genetic variation in offspring

Concept 13.3: Meiosis reduces the number of chromosome sets from diploid to haploid

- Like mitosis, meiosis is preceded by the replication of chromosomes
- Meiosis takes place in two sets of cell divisions, called **meiosis I** and **meiosis II**
- The two cell divisions result in four daughter cells, rather than the two daughter cells in mitosis
- Each daughter cell has only half as many chromosomes as the parent cell

The Stages of Meiosis

- After chromosomes duplicate, two divisions follow
 - Meiosis I (reductional division): homologs pair up and separate, resulting in two haploid daughter cells with replicated chromosomes
 - Meiosis II (equational division) sister chromatids separate
- The result is four haploid daughter cells with unreplicated chromosomes
- Meiosis I is preceded by interphase, when the chromosomes are duplicated to form sister chromatids
- The sister chromatids are genetically identical and joined at the centromere
- The single centrosome replicates, forming two centrosomes
- Division in meiosis I occurs in four phases
 - Prophase I
 - Metaphase I
 - Anaphase I
 - Telophase I and cytokinesis

Prophase I

- Prophase I typically occupies more than 90% of the time required for meiosis
- Chromosomes begin to condense
- In **synapsis**, homologous chromosomes loosely pair up, aligned gene by gene
- In **crossing over**, nonsister chromatids exchange DNA segments
- Each pair of chromosomes forms a tetrad, a group of four chromatids
- Each tetrad usually has one or more **chiasmata**, X-shaped regions where crossing over occurred

Anaphase I

- In anaphase I, pairs of homologous chromosomes separate
- One chromosome moves toward each pole, guided by the spindle apparatus
- Sister chromatids remain attached at the centromere and move as one unit toward the pole

Telophase I and Cytokinesis

- In the beginning of telophase I, each half of the cell has a haploid set of chromosomes; each chromosome still consists of two sister chromatids
- Cytokinesis usually occurs simultaneously, forming two haploid daughter cells
- In animal cells, a cleavage furrow forms; in plant cells, a cell plate forms
- No chromosome replication occurs between the end of meiosis I and the beginning of meiosis II because the chromosomes are already replicated

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- Division in meiosis II also occurs in four phases
 - Prophase II
 - Metaphase II
 - Anaphase II
 - Telophase II and cytokinesis
- Meiosis II is very similar to mitosis

Prophase II

- In prophase II, a spindle apparatus forms
- In late prophase II, chromosomes (each still composed of two chromatids) move toward the metaphase plate

Metaphase II

- In metaphase II, the sister chromatids are arranged at the metaphase plate
- Because of crossing over in meiosis I, the two sister chromatids of each chromosome are no longer genetically identical
- The kinetochores of sister chromatids attach to microtubules extending from opposite poles

Anaphase II

- In anaphase II, the sister chromatids separate
- The sister chromatids of each chromosome now move as two newly individual chromosomes toward opposite poles

Telophase II and Cytokinesis

- In telophase II, the chromosomes arrive at opposite poles
- Nuclei form, and the chromosomes begin decondensing
- Cytokinesis separates the cytoplasm
- At the end of meiosis, there are four daughter cells, each with a haploid set of unreplicated chromosomes
- Each daughter cell is genetically distinct from the others and from the parent cell

A Comparison of Mitosis and Meiosis

- Mitosis conserves the number of chromosome sets, producing cells that are genetically identical to the parent cell
- Meiosis reduces the number of chromosome sets from two (diploid) to one (haploid), producing cells that differ genetically from each other and from the parent cell
- Three events are unique to meiosis, and all three occur in meiosis I
 - Synapsis and crossing over in prophase I: Homologous chromosomes physically connect and exchange genetic information
 - At the metaphase plate, there are paired homologous chromosomes (tetrads), instead of individual replicated chromosomes
 - At anaphase I, it is homologous chromosomes, instead of sister chromatids, that separate
- Sister chromatid cohesion allows sister chromatids of a single chromosome to stay together through meiosis I

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- Protein complexes called cohesins are responsible for this cohesion
- In mitosis, cohesins are cleaved at the end of metaphase
- In meiosis, cohesins are cleaved along the chromosome arms in anaphase I (separation of homologs) and at the centromeres in anaphase II (separation of sister chromatids)

Concept 13.4: Genetic variation produced in sexual life cycles contributes to evolution

- Mutations (changes in an organism's DNA) are the original source of genetic diversity
- Mutations create different versions of genes called alleles
- Reshuffling of alleles during sexual reproduction produces genetic variation

Origins of Genetic Variation Among Offspring

- The behavior of chromosomes during meiosis and fertilization is responsible for most of the variation that arises in each generation
- Three mechanisms contribute to genetic variation
 - Independent assortment of chromosomes
 - Crossing over
 - Random fertilization

Independent Assortment of Chromosomes

- Homologous pairs of chromosomes orient randomly at metaphase I of meiosis
- In independent assortment, each pair of chromosomes sorts maternal and paternal homologs into daughter cells independently of the other pairs
- The number of combinations possible when chromosomes assort independently into gametes is 2^n , where n is the haploid number
- For humans ($n = 23$), there are more than 8 million (2^{23}) possible combinations of chromosomes

Crossing Over

- Crossing over produces **recombinant chromosomes**, which combine DNA 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
- Crossing over contributes to genetic variation by combining DNA from two parents into a single chromosome

Random Fertilization

- Random fertilization adds to genetic variation because any sperm can fuse with any ovum (unfertilized egg)
- The fusion of two gametes (each with 8.4 million possible chromosome combinations from independent assortment) produces a zygote with any of about 70 trillion diploid combinations
- Crossing over adds even more variation

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- Each zygote has a unique genetic identity

The Evolutionary Significance of Genetic Variation Within Populations

- Natural selection results in the accumulation of genetic variations favored by the environment
- Sexual reproduction contributes to the genetic variation in a population, which originates from mutations