# Meiosis Notes

I. Each parent donates genes to their offspring via sexual reproduction. The genes combine to give different but similar looking offspring.

A. In humans, each **somatic cell** (all cells other than sperm or ovum) has 46 chromosomes, consisting of 23 homologous pairs.

1. Any cell with two sets of chromosomes is called a **diploid cell** and has a diploid number of chromosomes, abbreviated as 2n.

B. Each parent donates one chromosome to each of the 23 homologous pairs. *I.e.*, half of an individual's chromosomes come from the female parent while half come from the male parent.

C. Reproductive cells are called **gametes** (sperm and eggs) and have only one set of chromosomes ― 22 autosomes and an X (in an ovum) and 22 autosomes and an X or a Y (in a sperm cell). Remember, autosomes are chromosomes other than the sex chromosomes.

1. The number of chromosomes in a single set is represented by *n*.
2. Such cells are called **haploid**.

D. For humans, the haploid number of chromosomes is 23 ($n = 23$), and the diploid number is 46 ($2n = 46$).

E. Images of the 46 human chromosomes can be arranged in pairs in order of size to produce a **karyotype** display.

1. The two chromosomes comprising a pair have the same length, centromere position, and staining pattern.
2. These **homologous chromosome** pairs carry genes that control the same inherited characters.
3. Homologous chromosomes are the same length and carry the same genes in the same location. Those genes could be different versions. *E.g.*, imagine the homologous chromosomes carry the eye color gene but one produces blue eyes while the other produces brown.
4. The exception is the sex chromosomes. For these, females have a homologous pair (XX) while males do not (XY).
5. The other 22 pairs of chromosomes are called autosomes.

II. Meiosis reduces the number of chromosomes in a cell by half.

A. If one somatic cell is fertilized by another, the resulting zygote would contain twice the number of chromosomes. *I.e.*, the chromosome number would double each generation.

B. For this reason, the chromosome number must be reduced during the production of gametes. This way, two haploid gametes fuse to form a diploid zygote.

C. Meiosis is quite similar to mitosis except there are two consecutive cell divisions, **meiosis I** and **meiosis II,** resulting in four daughter cells.

1. The first division, meiosis I, separates homologous chromosomes.
2. The second, meiosis II, separates sister chromatids.

D. Meiosis I is preceded by **interphase,** in which the chromosomes are replicated to form sister chromatids.

1. These are genetically identical and joined at the centromere.

E. Meiosis I consists of four phases: prophase I, metaphase I, anaphase I, and telophase I.

1. Prophase I typically occupies more than 90% of the time required for meiosis.
   a. During prophase I, the chromosomes begin to condense.
   b. Homologous chromosomes loosely pair up along their length, precisely aligned gene for gene.

(1) In **synapsis** , homologous chromosomes line up beside one another.
(2) The group of four chromatids is called a **tetrad**.
(3) During synapsis, the arms of chromosomes in a homologous pair become intertwined. Pieces of the homologous chromosomes break off and switch places. This phenomenon is called **crossing over**.

2. At **metaphase I**, the tetrads are all arranged at the metaphase plate, with one chromosome facing each pole.
3. In **anaphase I**, the homologous chromosomes separate.
4. In **telophase I**, movement of homologous chromosomes continues until there is a haploid set at each pole.
   a. Each chromosome consists of two sister chromatids.
5. **Cytokinesis** usually occurs simultaneously, by the same mechanisms as mitosis.
   a. In animal cells, a cleavage furrow forms. In plant cells, a cell plate forms.
6. No chromosome replication occurs between the end of meiosis I and the beginning of meiosis II, as the chromosomes are already replicated.

F. Meiosis II is very similar to mitosis.
1. During **prophase II**, spindle fibers attach to each sister chromatid.
2. At **metaphase II**, the sister chromatids are arranged at the metaphase plate.
   a. Because of crossing over in meiosis I, the two sister chromatids of each chromosome are no longer genetically identical.
   b. At **anaphase II**, the centomeres of sister chromatids separate and the two now individual chromosomes travel toward opposite poles.
3. In **telophase II**, the chromosomes arrive at opposite poles.
   a. Nuclei form around the chromosomes, which begin expanding, and cytokinesis separates the cytoplasm.

III. Events in meiosis contribute to genetic diversity.
A. Genetic diversity contributes to evolutionary change.
1. Differences in genes arise by mutation.
2. If an offspring inherits a combination of genes that gives it a survival advantage, it is better able to survive and pass on its genes. This means the chance that the combination is passed on increases.
3. As a result, there is an accumulation of favorable characteristics.

B. These different versions are reshuffled during meiosis and fertilization, producing offspring with their own unique set of traits.

C. **Independent assortment of chromosomes** contributes to genetic variability due to the random orientation of homologous pairs of chromosomes at the metaphase plate during meiosis I.
1. 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.
2. Each homologous pair of chromosomes segregates independently of the other homologous pairs during metaphase I.
3. The number of combinations possible when chromosomes assort independently into gametes is $2^n$, where n is the haploid number of the organism.
   a. For humans with $n = 23$, there are $2^{23}$, or more than 8 million possible combinations of chromosomes.

D. **Crossing over** produces **recombinant chromosomes,** which combine genes inherited from each parent.
1. For humans, this occurs an average of one to three times per chromosome pair.

E. **Random fertilization** adds to the genetic variation arising from meiosis.
   1. Any sperm can fuse with any egg.
   2. The ovum is one of more than 8 million possible chromosome combinations.
   3. The successful sperm is one of more than 8 million possibilities.
   4. The resulting zygote could contain any one of more than 70 trillion possible combinations of chromosomes.Evolution - if offspring inherit a combination that is more successful they increase the chances of passing it to offspring. The result is an accumulation of positive traits.

IV. Alterations of chromosome number cause some genetic disorders.
   A. Tests are available to determine *in utero* if a child has a particular disorder.
      1. **Amniocentesis** can be used from the 15$^{th}$ to the 20$^{th}$ week of pregnancy.
         a. Amniotic fluid is withdrawn from the amniotic sac and checked for the presence of disease-causing alleles.
         b. Some disorders can be detected by the presence of certain chemicals in the fluid while fetal cells present in the fluid can be grown to be used for karyotyping to identify chromosomal abnormalities.
      2. A second technique, **chorionic villus sampling (CVS)** allows faster karyotyping and can be performed as early as the 10-12 weeks pregnant (although it has been used as early as the 8$^{th}$ week of pregnancy).
         a. A small amount of fetal tissue is removed from the placenta and the cells are used for karyotyping.
         b. The risk might be higher for this procedure but it may be chosen because it can be done earlier in pregnancy.
      3. Other techniques, **ultrasound** and **fetoscopy**, can be used to assess fetal health visually *in utero.*
      4. These techniques can cause complications which lead to loss of the pregnancy in between 0.5-1% of cases and so are only used in cases for which the risk of a genetic disorder or other type of birth defect is relatively high.
      5. If fetal tests reveal a serious disorder, the parents face the difficult choice of terminating the pregnancy or preparing to care for a child with a genetic disorder.
      6. Some disorders can be detected at birth by test that are routinely performed on newborns
   B. The most common technique for diagnosing chromosomal abnormalities (including nondisjunction disorders) is the **karyotype**.
      1. To prepare a karyotype, fetal cells must be harvested and cultured *in vitro*.
      2. The chromosomes are removed and arranged in pairs and photographed.
      3. Abnormalities in chromosome number, shape, or size can be detected this way.
   C. **Nondisjunction** occurs when problems with the meiotic spindle cause abnormal separation of the chromosomes into daughter cells.
      1. This may occur if tetrad chromosomes do not separate properly during meiosis I.
      2. Alternatively, sister chromatids may fail to separate during meiosis II.
      3. As a consequence of nondisjunction, one gamete receives two of the same type of chromosome, and another gamete receives no copy.
      4. Offspring resulting from fertilization of a normal gamete with one produced by nondisjunction will have an abnormal chromosome number, a condition known as **aneuploidy.**
         a. **Trisomic** cells have three copies of a particular chromosome type and have $2n + 1$ total chromosomes.

b. **Monosomic** cells have only one copy of a particular chromosome type and have $2n - 1$ chromosomes.

5. If this happens early in development, because of an error in the mitotic spindle, the aneuploid condition will be passed along by mitosis to a large number of cells.
   a. This is likely to have a substantial effect on the organism.
   b. If the organism survives, aneuploidy typically leads to a distinct phenotype.
6. Although the frequency of aneuploid zygotes may be quite high in humans, most of these alterations are so disastrous to development that the embryos are spontaneously aborted long before birth.
   a. **Down syndrome** is caused by having three copies of chromosome 21 or trisomy 21.
      (1) Individuals with Down syndrome have characteristic facial features, short stature, heart defects, susceptibility to respiratory infection, mental retardation, and increased risk of developing leukemia and Alzheimer's disease.
      (2) Most are sexually underdeveloped and sterile.
   b. Trisomy 13 results in **Patau syndrome**.
      (1) Affected individuals have serious eye, brain, and circulatory defects and a very short life span - rarely more than one year.
   c. **Edward's syndrome** is caused by trisomy 18.
      (1) The syndrome affects most body organs and life span is usually less than 10 weeks.
   d. An XXy male has **Klinefelter's syndrome**.
      (1) These individuals have male sex organs, but the testes are abnormally small and fail to descend. Individuals are sterile.
      (2) Although the extra X is inactivated, some breast enlargement and other female characteristics are common.
      (3) Affected individuals have normal intelligence.
   e. Some research has suggested that males with an extra Y chromosome (Xyy) tend to be slighty taller than average.
   f. Trisomy X (XXX) females are indistinguishable from normal females..
   g. Monosomy X or **Turner syndrome** (X0) occurs once in every 5,000 births.
      (1) This is the only known viable monosomy in humans.
      (2) X0 individuals are phenotypically female but are sterile because their sex organs do not mature.
      (3) When provided with estrogen replacement therapy, girls with Turner syndrome develop secondary sex characteristics.
      (4) Most are of normal intelligence.
7. Chromosome deletions can also cause severe disorders.
   a. One syndrome, *cri du chat*, results from a specific deletion in chromosome 5.
      (1) These individuals are mentally retarded, have small heads with unusual facial features, and have a cry like the mewing of a distressed cat.
      (2) This syndrome is fatal in infancy or early childhood.
8. Chromosomal translocations between nonhomologous chromosomes are also associated with human disorders.
   a. Chromosomal translocations have been implicated in certain cancers, including *chronic myelogenous leukemia (CML).*
   b. CML occurs when a large fragment of chromosome 22 switches places with a small fragment from the tip of chromosome 9.

D. It is believed that many cases of mental retardation are linked to chromosomal defects.

E. Most certainly nondisjunction occurs in other chromosomes but the consequences are lethal.