AP Biology

Unit 5

Student Notes

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Important Ideas/Enduring Understandings for this unit.

- Heritable information provides for the continuity of life.
- Organisms are linked by line of descent from common ancestry.
- Naturally occurring diversity among and between components within biological systems affects interactions with the environment.

Chromosomes, Meiosis, and Sexual Reproduction

Important Terms

Heredity

Heredity refers to the transmission of traits from one generation to the next by **INHERITING DNA** from a single parent (asexual reproduction) or from two parents (sexual reproduction). The genetic information transmitted via the DNA provides for the continuity of life across generations. Major features of the genetic code and many of the core metabolic pathways regulated by genes are conserved across all living things. All life forms possess DNA, RNA, and ribosomes and carry out the processes of DNA replication, transcription, and translation (protein synthesis) in very similar ways. This high level of conservation supports the concept of common ancestry for all organisms.

Chromosome

Chromosomes are the packaged and organized structures of DNA found in cells. Chromosomes consist of chains of linked genes and associated proteins. The chromosomal basis of inheritance can be used to explain the pattern of transmission of genes from parent(s) to offspring.

A **eukaryotic chromosome** consists of a single molecule of DNA wrapped around proteins called **histones**. The histones function to package and organize the DNA. They also play an important role in gene regulation.

Eukaryotic chromosomes are only visible (with a microscope) just before and during **cell division**. During other parts of the cell cell cycle, the DNA is present in an a less condensed form known as **chromatin**. In this form, the DNA can be transcribed and replicated.

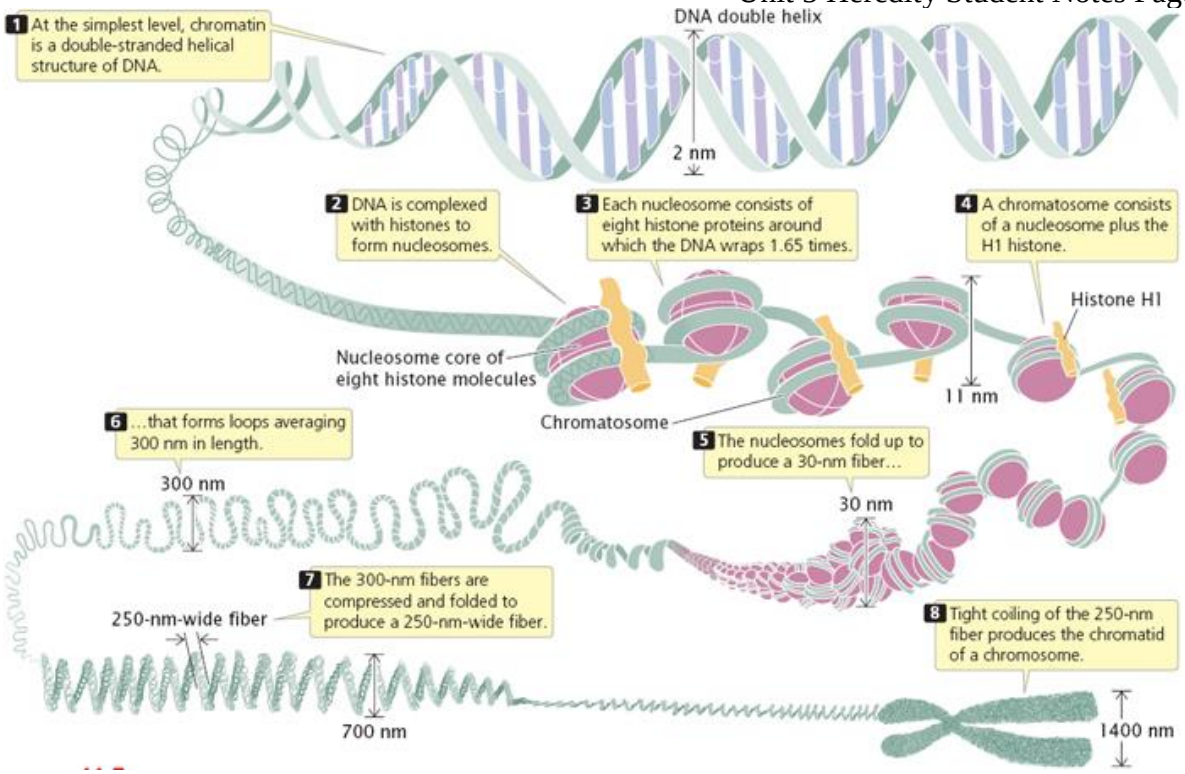
Each chromosome contains several genes and the other DNA sequences involved in the regulation of the genes. Genes that are located on the same chromosome are said to be **linked.**

Eukaryotic chromosomes are said to have a **linear shape**.

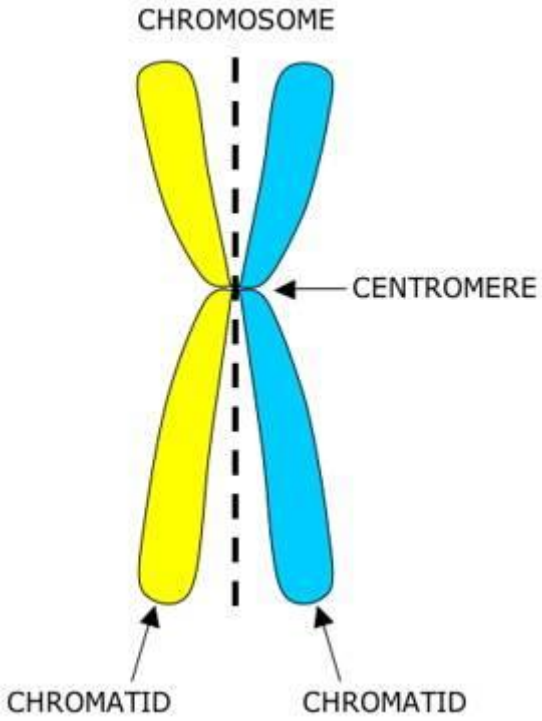
After replication, eukaryotic chromosomes often have an "X-shaped" appearance. These structures are more correctly referred to as **bivalent chromosomes**. They consist of two copies of the original chromosome attached at a central point known as the **centromere**. While connected the two copies are referred to as **chromatids.**

Chromosomes that are not involved in gender determination are called **autosomes**. Human cells contain 44 autosomes.

Chromosomes that are involved in the process of gender determination are called **sex chromosomes.** Human females have two X sex chromosomes, while human males have one X and one Y sex chromosome.


1 At the simplest level, chromatin is a double-stranded helical structure of DNA.
DNA double helix
2 nm
2 DNA is complexed with histones to form nucleosomes.
3 Each nucleosome consists of eight histone proteins around which the DNA wraps 1.65 times.
4 A chromatosome consists of a nucleosome plus the H1 histone.
Histone H1
Nucleosome core of eight histone molecules
11 nm
Chromatosome
5 The nucleosomes fold up to produce a 30-nm fiber…
30 nm
6 …that forms loops averaging 300 nm in length.
300 nm
7 The 300-nm fibers are compressed and folded to produce a 250-nm-wide fiber.
250-nm-wide fiber
8 Tight coiling of the 250-nm fiber produces the chromatid of a chromosome.
700 nm
1400 nm

BIVALENT


CHROMOSOME
CENTROMERE
CHROMATID
CHROMATID

A **prokaryotic chromosome** is circular and resides in a cell region called the **nucleoid**. Prokaryotes typically have only one chromosome per cell. The types of proteins found in prokaryotic chromosomes, known as the nucleoid-associated proteins, differ from the histone proteins that appear in eukaryotic chromosomes and cause the prokaryotic chromosomes to form looped structures. In addition to the single, circular chromosome, many prokaryotes also contain plasmids. A **plasmid** is a small, circular, double-stranded DNA molecule that is distinct from a cell's chromosomal DNA. **Plasmids** naturally exist in bacterial cells, and they also occur in some eukaryotes. Often, the genes carried in **plasmids** provide bacteria with genetic advantages, such as antibiotic resistance. Plasmids can be exchanged between bacterial cells during the process of **conjugation**. During conjugation the bacterial cells attach to each other and exchange plasmids. This is an important process for increasing genetic variation within the population.

Gene

A gene is a segment of DNA which codes for a **single polypeptide or protein**. Each chromosome consists of a chain of several genes and the accompanying regulatory sequences and associated proteins.

Genome

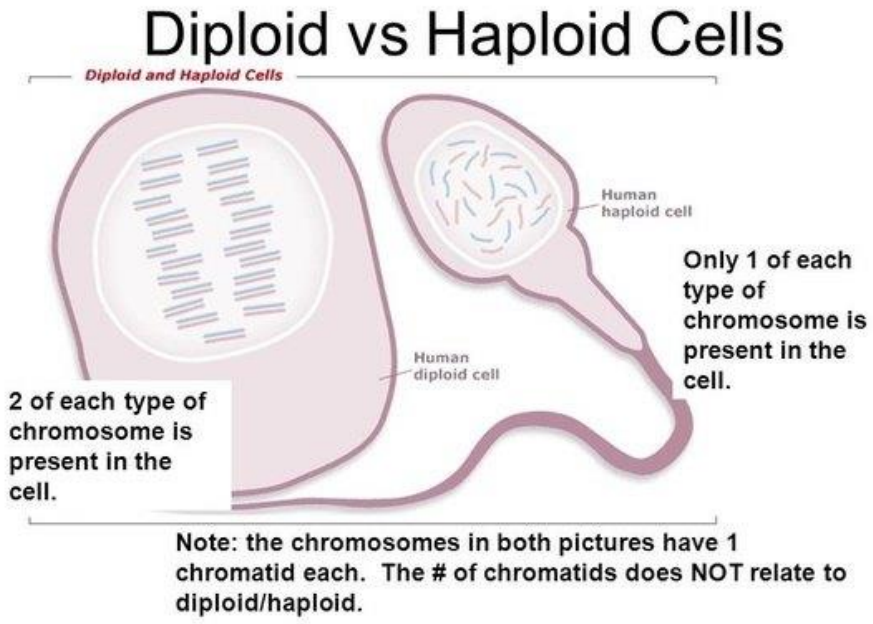
A **genome** is an organism's complete set of DNA, including all of its genes. Each **genome** contains all of the information needed to build and maintain that organism. In humans, a copy of the entire **genome**—more than 3 billion DNA base pairs—is contained in all cells that have a nucleus.

Haploid

Haploid refers to cells that have only one copy of each type of chromosome. Haploid cells are sometimes referred to as **N or 1N cells**. In human cells, the haploid number is 23. The only haploid cells found in the human body are the gametes or sex cells.

Diploid

Diploid cells have two copies of each type of chromosome. Diploid cells are sometimes referred to as 2N cells. In human cells, the diploid number is 46. All of the human body's somatic cells are diploid.



Locus

The **location** of a gene on a chromosome.

Types of Reproduction

Asexual Reproduction

Asexual reproduction involves only ONE parent. During the process, the parent produces clones which are genetically identical to itself. The benefits of asexual reproduction are that the process can occur rapidly, the process requires less energy than sexual reproduction, and that only one organism is required to carry out the process. Examples of asexual reproduction include binary fission in bacteria, budding in yeasts and hydras, and vegetative propagation in some plants.

The major disadvantage of asexual reproduction is that it produces identical organisms. Since there is no variation, the ability of the population to adapt/evolve is severely limited. Asexual reproduction works well in stable environments (to which the organism is well adapted), but does not work well in changing environments to which populations need to be able to adapt.

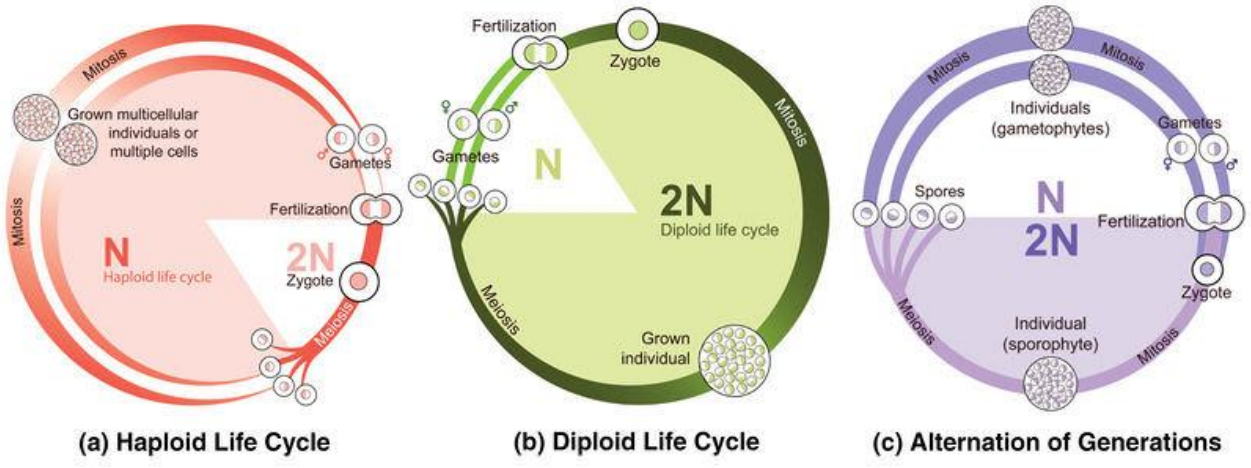
Sexual Reproduction

Sexual reproduction involves the combining of DNA from two parents to create an offspring. During fertilization, two haploid games fuse together to form a zygote. Fertilization restores the diploid number of chromosomes and maintains in from generation to generation. The major benefit of the sexual reproduction is that it creates genetic diversity/variation within the population by combining the DNA from two different individuals to form a new organism with new allele combinations. This variation allows the population to adapt/evolve with a changing environment. The major disadvantages of sexual reproduction are that two parents of opposite sexes are required (this may be nearly impossible for endangered species to be able to do), the process takes much longer than asexual reproduction, and the process requires more energy than asexual reproduction. Most animals and plants reproduce sexually. Some protists and fungi also utilize sexual reproduction.

	Sexual Reproduction	Asexual Reproduction
Advantages	• High Genetic Variability • Facilitates adaptation • "Speeds" up evolution	• Saves energy • Courtship is a non-issue • Greatest increase in fitness for each individual
Disadvantages	• Energy Costly • Courtship is time/resource consuming • Usually sacrifices the fitness of one sex to the other.	• Low Genetic Variability • Adaptation to environment is difficult • "Retards" evolution

Types of Life Cycle

Most living things follow one of three possible types of life cycles, based on the amount of DNA within their cells.



(a) Haploid Life Cycle (b) Diploid Life Cycle (c) Alternation of Generations

Haploid Majority

Organisms with a haploid life cycle spend the majority of their lives as haploid cells. When two of the haploid cells fuse, they form a diploid zygote. The zygote quickly undergoes meiosis to produce more haploid cells that repeat the life cycle.
This haploid majority life cycle occurs mostly in Fungi and Protists.
The fusion of the haploid cells and the accompanying meiosis event, functions to create genetic diversity/variation within the population.

Diploid Majority

Organisms with a diploid life cycle spend the majority of their lives as multicellular organisms composed of diploid cells. In these organisms, diploid germ cells undergo meiosis to create haploid gametes/sex cells. During fertilization, a haploid egg fuses with a haploid sperm to create a diploid zygote. The zygote undergoes repeated rounds of mitosis to create a multicellular organism composed of diploid cells. Most animals exhibit a diploid majority life cycle.

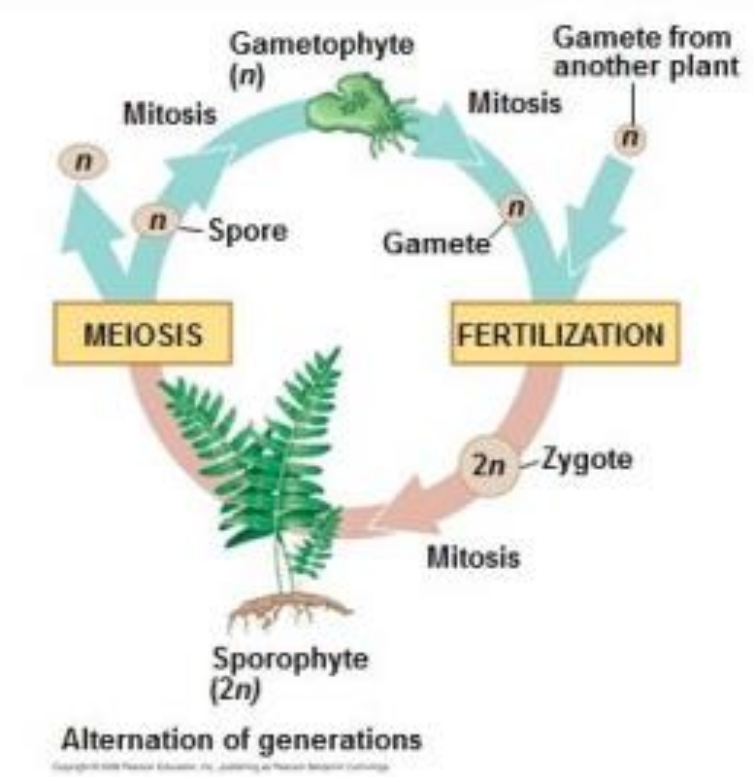
Alternation of Generations

Organisms which exhibit an alternation of generations alternate between a haploid sexual phase (the gametophyte) and a diploid asexual phase (the sporophyte) of the life cycle.

The **gametophyte generation** begins with a **spore** produced by meiosis. The spore is haploid, and all of the cells derived from it (by mitosis) are also haploid. In due course, this multicellular structure produces gametes — by **mitosis** — and the fusion of two of these gametes then produces the diploid **sporophyte generation**.

The sporophyte generation starts as a diploid zygote. Eventually, certain cells within the organism undergo meiosis, forming spores. Each spore is capable of developing into a new haploid gametophyte. Most plants exhibit an alternation of generations.

In bryophytes, such as mosses and liverworts, the gametophyte is the dominant life phase, whereas in angiosperms and gymnosperms the sporophyte is dominant. The haploid phase is also dominant among fungi.



Meiosis

Meiosis is a specialized type of **nuclear division (in sexually reproducing diploid organisms)** that reduces the chromosome number by half, creating four haploid cells, each genetically distinct from the parent cell that gave rise to them. The haploid cells, after undergoing a period of maturation, become gametes.
The two main roles of meiosis are to create haploid gametes and to increase genetic diversity/variation within the gametes.

In multicellular animals, meiosis occurs only in the **sex organs/gonads**. The only cells capable of doing meiosis are **specialized, diploid germ cells** found in the gonads.

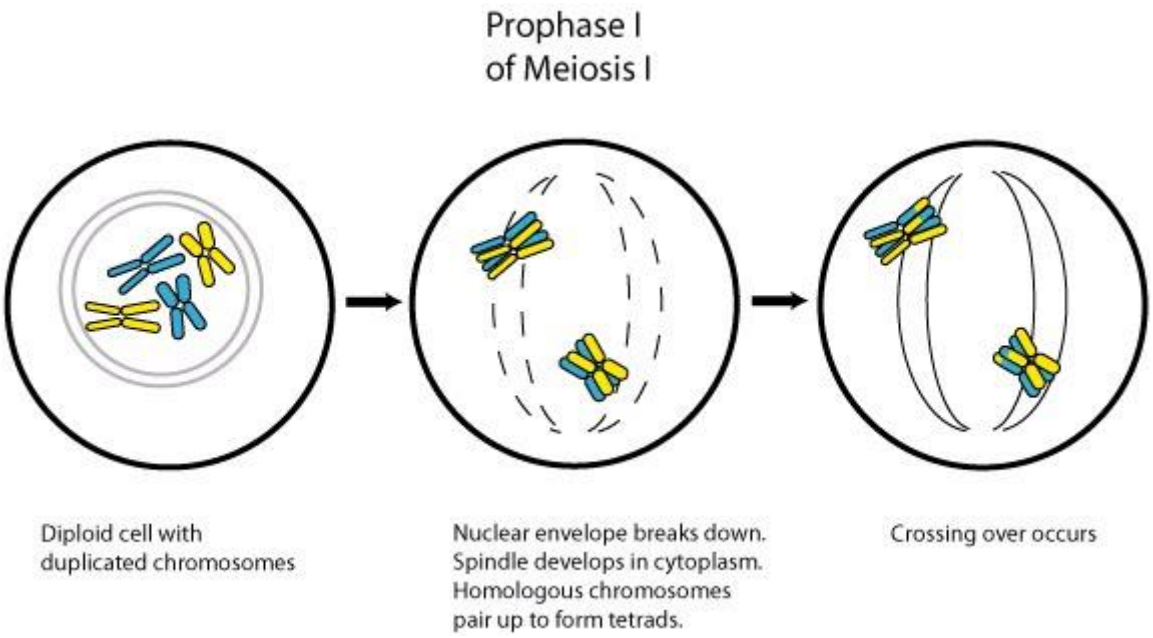
Before meiosis begins, the DNA is replicated during the S phase of interphase.

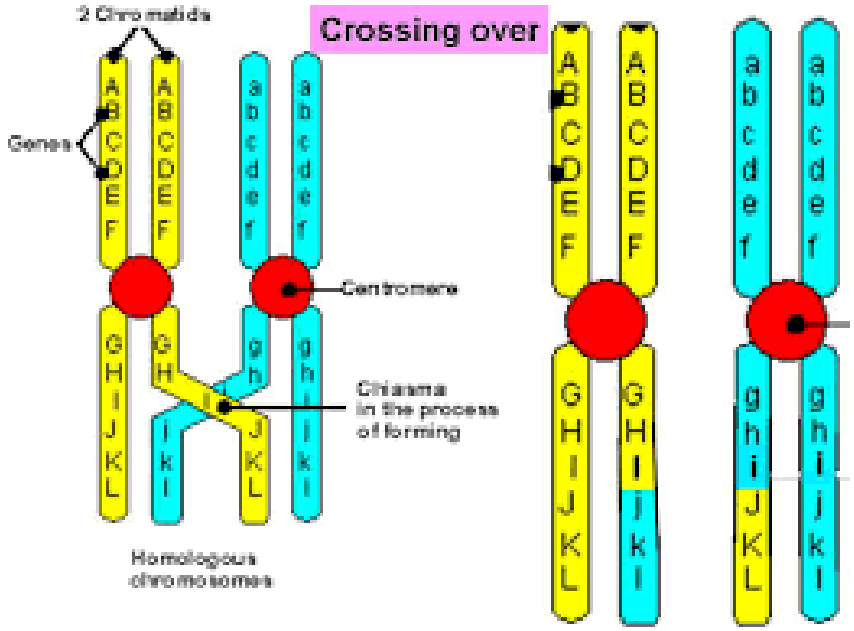
Meiosis involves two rounds of a sequential series of steps. These rounds are referred to as Meiosis I and Meiosis II.

Stages of Meiosis
Meiosis I

Prophase I
Meiosis begins with Prophase I. During Prophase I, the cell's chromatin coils and condenses to form bivalent chromosomes. The nucleolus disappears. The centrosomes form the spindle fibers and the nuclear envelope disintegrates. Chromosomes of the same type/number (**homologous chromosomes**) pair up and join together during the process of **synapsis** to form tetrads (groups of 4 homologous chromatids). The members of each pair contain the same genes in the same order on the chromosome, but may contain different alleles or versions of the genes. After synapsis, the homologous chromosomes undergo the process of **crossing over** in which they physically exchange segments. This is an example of

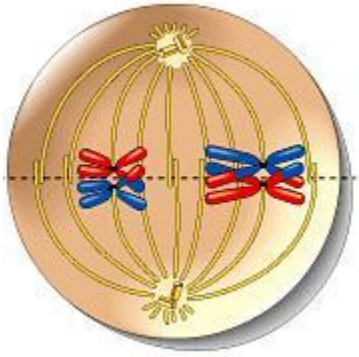
genetic recombination. The process is an important source of genetic variation/diversity in the gametes that result at the end of meiosis.





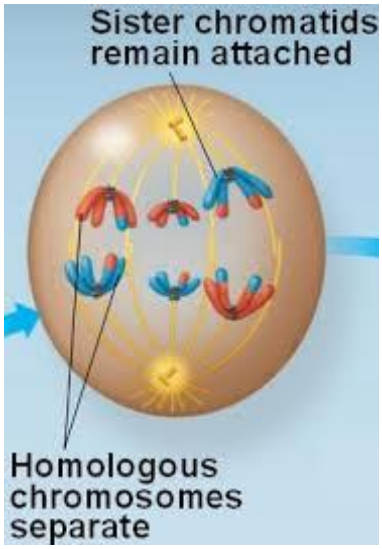
Metaphase I

During Metaphase I, the two members of each homologous pair of bivalent chromosomes align on each side of the **metaphase plate** (middle of the cell). Each chromosome also attaches to the spindle fibers at a point on the centromere known as the **kinetochore**. During the process of lining up, the paternal and maternal members of each homologous pair randomly line up across from each other on either side of the metaphase plate. The pattern in which each pair arranges itself is completely independent of the other homologous pairs. This process of random alignment, known as **independent assortment**, can create up to 2^{23} (8,388,608) possible alignments in human germ cells. Since the two rows of chromosomes will eventually end up in different gametes, the process of independent assortment gives each human the ability to create over 8 million genetically distinct types of gametes**. I**ndependent assortment increases genetic variation by allowing daughter cells to each ***randomly*** receive a different proportion of paternal and maternal chromosomes.



In the diagram above, red chromosomes represent maternal chromosomes and blue ones represent paternal chromosomes. During independent assortment, both red, maternal chromosomes could face the top pole and both blue chromosomes could face the bottom pole or both blue chromosomes could face the top pole and both red could face the bottom or one red on the left and one blue on the right could face the top or one blue on the left and one red on the right could face the top. It turns out the total possible combinations of alignments due to independent assortment can be found by raising 2 to the number of homologous pairs contained within the cell. **Number of combinations=$2^{\text{number of homologous pairs}}$**
For example, a cell with 6 homologous pairs of chromosomes could independently assort into 2^6 or 64 different arrangements.

Anaphase I
During Anaphase I, one member of each homologous pair of chromosomes is pulled toward each pole of the cell by the microtubules of the spindle apparatus. This process is called **Segregation**. The individual chromatids of each bivalent chromosome are not separated as they are during the anaphase stage of mitosis. Anaphase I separates the members of the homologous pairs from each other. This process of separation ensures that each gamete receives a haploid (1N) set of chromosomes comprised of a mixture of chromosomes from both parents.



Telophase I
During Telophase I, the homologous chromosome pairs reach the poles of the cell, the spindle fibers disappear, nuclear envelopes form around each set of chromosomes, and **cytokinesis** follows to produce two cells. The two new cells are haploid and have half the number of chromosomes as the original germ cell. It is important to note that we count chromosomes by counting the number of centromeres present. Even though the chromosomes at the end of telophase I are bivalent, each bivalent chromosome has one centromere and only counts as a single chromosome.

Telophase I

- Two daughter cells are formed with each one containing only one chromosome of the homologous pair.
- The daughter cells are now haploid.

Meiosis I is often referred to as **reductive division**, because it halves the number of chromosomes found in the two cells which result from the process.

To summarize, Meiosis I begins with 1 diploid germ cell and ends with 2 haploid cells which both possess bivalent chromosomes.

Interkinesis

Interkinesis is a short interphase-like period that occurs between Meiosis 1 and Meiosis 2. **No DNA replication** occurs during interkinesis, however it does occur during the interphase I stage that occurs before meiosis begins. During interkinesis, the single spindle of the first meiotic division disassembles and the microtubules reassemble into two new spindles for the second meiotic division.

Meiosis 2

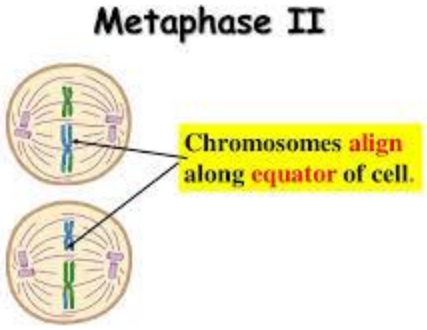
Prophase II

The nuclear envelopes and the nucleoli of **each** of the cells produced during Meiosis 1 disintegrate during prophase II. The chromosomes condense and the centrosomes replicate and move towards opposite poles. Spindle fibers grow outward from the centrosomes. Prophase II is almost identical to the Prophase stage of mitosis. No synapsis or crossing over occur during this stage.

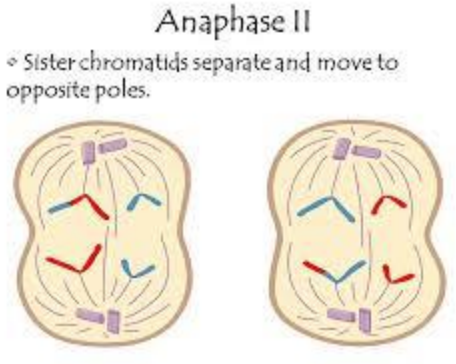
Prophase II

Metaphase II

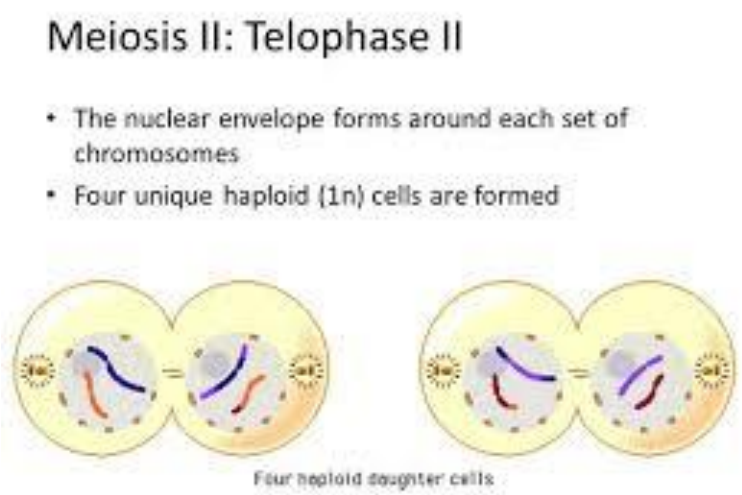
The chromosomes in both cells become arranged on the **metaphase plate**, much as the chromosomes do in mitosis, and are attached to the now fully formed spindle.



Anaphase II
During Anaphase II, the centromeres of each chromosome (in both cells) separate and the sister chromatids—now called individual chromosomes—move toward the opposite poles of the cells. Anaphase II is the process in which sister chromatids are separated from each other.

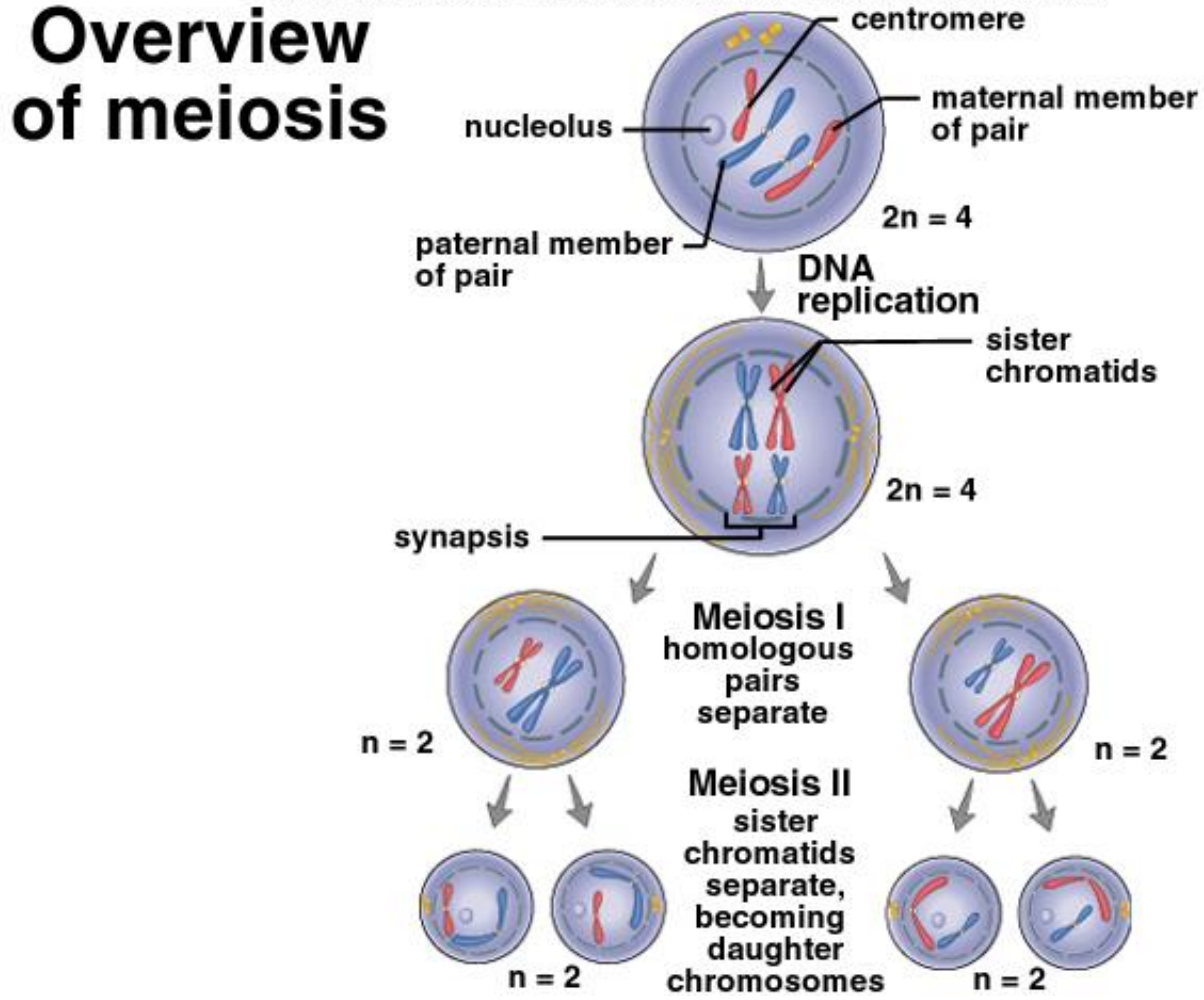


Telophase II
During Telophase II, a nuclear envelope forms around each set of chromosomes and **cytokinesis** occurs, producing four genetically different daughter cells, each with a haploid set of chromosomes.



Meiosis Summary

Meiosis is the first step in the process of gametogenesis or gamete creation. Meiosis happens only in the gonads and creates four haploid gametes (with single chromatid chromosomes) from each diploid germ cell that enters the process. Sexual reproduction in eukaryotes involving gamete formation/meiosis (including crossing over, the random/independent assortment of chromosomes during meiosis, and the subsequent fertilization of gametes) serves to increase the genetic variation within a population.

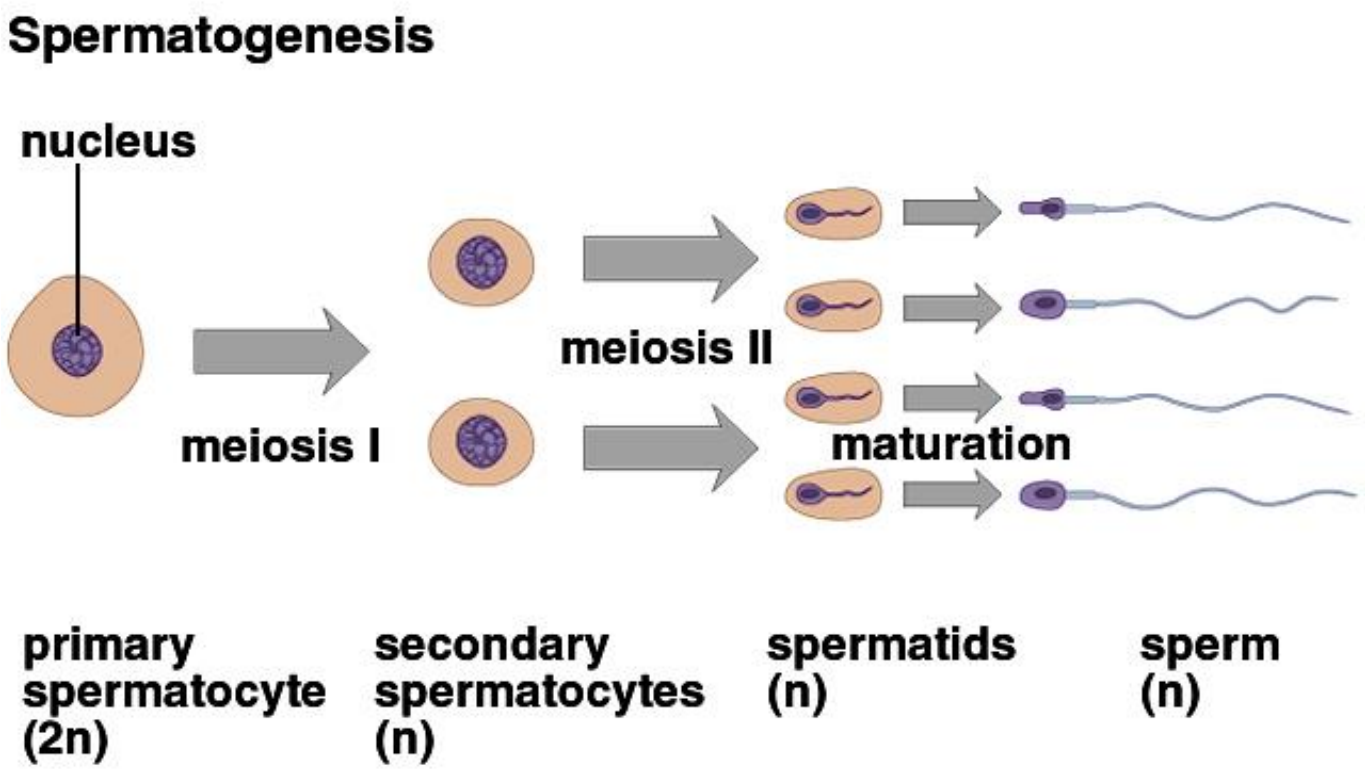


Gametogenesis

Gametogenesis is the process which creates gametes or sex cells. The process starts with meiosis. Meiosis is then followed by a period of maturation.

The process of sperm creation is known as **spermatogenesis.** Spermatogenesis begins when a diploid germ cell, known as a primary spermatocyte, undergoes the process of meiosis to create 4 haploid spermatids. The spermatids mature in the epididymis to become functional sperm. Men can continue to carry out the process of spermatogenesis from puberty until death.

Spermatogenesis and oogenesis

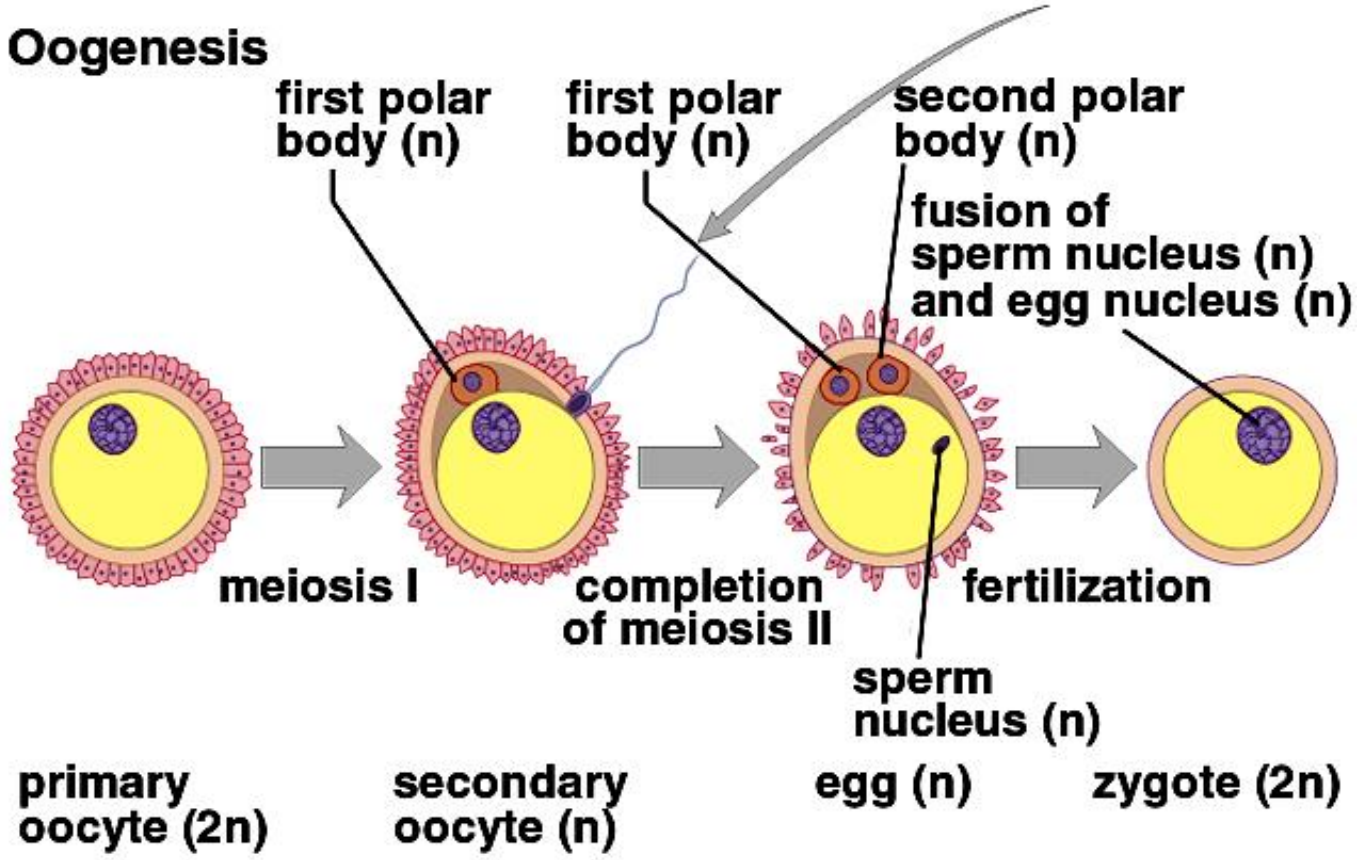


The process of egg creation is known as **oogenesis**. The diploid germ cells that have the potential to develop into ova are called oogonia. In humans, all of a female's oogonia that she will make in her lifetime are created when she's still a fetus and hasn't even been born yet. In fact, about one or two months before a baby girl is born, most of her approximately seven million oogonia die, and the remaining surviving oogonia enter meiosis I and become **primary oocytes**. These primary oocytes press the pause button on their development in prophase I, after they've replicated their genomes, but before they've made the first meiotic division. They stay arrested at this stage of development for over a decade until the girl begins her first menstrual cycle. Then, for about the next 30 to 45 years, on a monthly basis, some of the primary oocytes resume meiosis where they left off and complete the first meiotic division.

When the primary oocyte does finally complete its first meiotic division, it divides the chromosomes evenly, just as you would expect. However, **it does not divide its cytoplasm equally**. Almost all of the cytoplasm remains in one of the two daughter cells, which becomes a **secondary oocyte**. The other daughter cell, which gets half of the chromosomes but very little cytoplasm, is called a **polar body**. The polar body is not a functional oocyte, instead it **degenerates and dies.** The formation of a polar body allows the primary oocyte to reduce its genome by half and conserve most of its cytoplasm in the secondary oocyte. **This allows the egg cell to be large enough to store the nutrients and proteins needed for a developing offspring.**

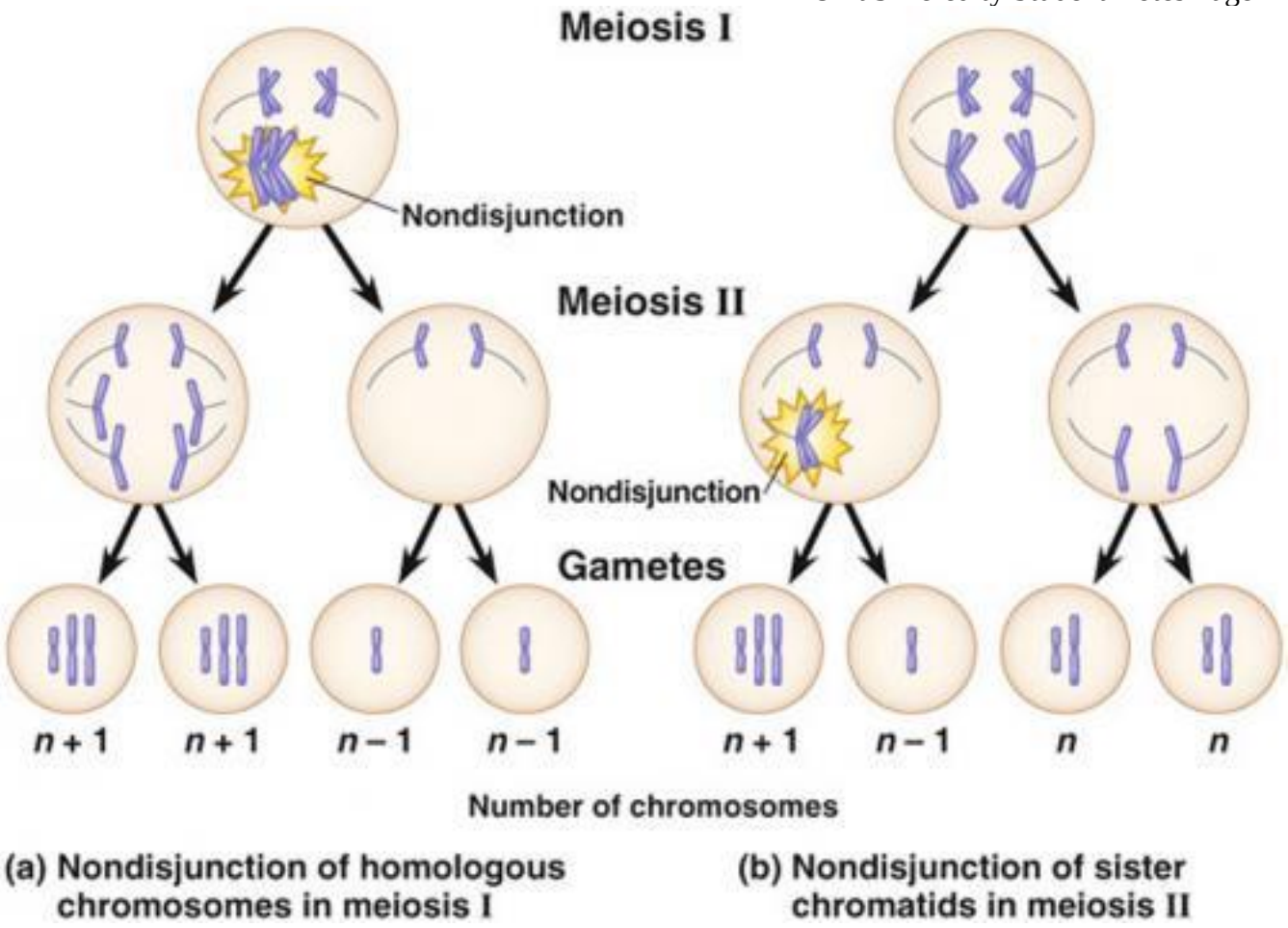
The secondary oocyte still has bivalent chromosomes, so if it's going to become a fully-functional ovum, it must undergo the Meiosis II. Meiosis II occurs only if the secondary oocyte is penetrated by a sperm cell. This division is also uneven, like the first one, with half of the chromosomes going to another very small degenerate polar body and half of the chromosomes being retained by the ovum (functional egg) along with almost all of the cytoplasm. In this way, the ovum achieves its haploid state while conserving as much cytoplasm as possible. **This means that during oogenesis, only one functional haploid egg is created from each diploid germ cell.**

Spermatogenesis and oogenesis



Nondisjunction

Nondisjunction is the failure of homologous chromosomes or sister chromatids to separate properly during cell division. There are three forms of nondisjunction: failure of a pair of homologous chromosomes to separate in meiosis I, failure of sister chromatids to separate during meiosis II, and failure of sister chromatids to separate during mitosis. Nondisjunction results in daughter cells with abnormal chromosome numbers, a condition called **aneuploidy**. When nondisjunction occurs during meiosis, it can result in gametes with the wrong number of chromosomes. If fertilized, the most common result is miscarriage. Most human embryos cannot survive with an abnormal chromosome number.



The letter "n" in the diagram included above indicates the normal chromosome number.

There are some conditions in which the aneuploid zygote survives. The table below describes some of the conditions which result.

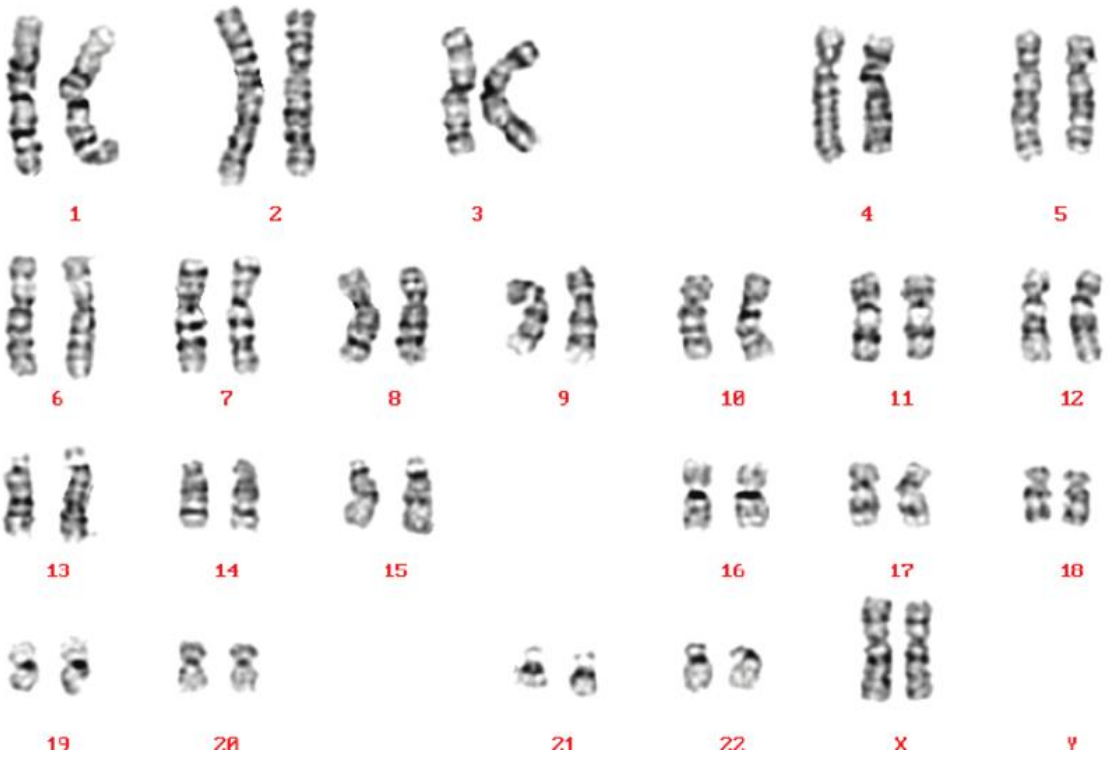
Nondisjunction syndrome frequencies

Syndrome	Description	Chromosomes	Incidences (newborns)
Down	Mental retardation; wide, flat face with upper eyelid fold, short stature; abnormal palm creases	Trisomy 21	1/800
Patau	Malformed internal organs, face, and head; extra digits; mental retardation	Trisomy 13	1/15,000
Edward	Malformed internal organs, face, and head; extreme muscle tone	Trisomy 18	1/6,000
Turner	Short stature; webbed neck; broad chest; no sexual maturity	XO	1/6,000
Klinefelter	Breast development possible; testes underdeveloped; no facial hair	XXY (or XXXY)	1/1,500
Triplo-X	Tall and thin with menstrual irregularities	XXX (or XXXX)	1/1,500
Jacob	Taller than average; persistent acne; speech and learning problems possible	XYY	1/1,000

b. From Robert F. Weaver and Philip W. Hedrick, *Genetics, 2nd ed.* Copyright 1992 WCB.

Many of these conditions can be diagnosed either before or right after birth by the analysis of a **karyotype**. A karyotype is simply a picture of a person's chromosomes. In order to get this picture, the chromosomes are isolated, stained, and examined under the microscope. Most often, this is done using the chromosomes in the white blood cells. A picture of the chromosomes is taken through the microscope. Then, the picture of the chromosomes is cut up and the chromosomes are arranged by size into homologous pairs. The chromosomes are lined up from largest to smallest. **The autosomes are displayed are displayed as pairs 1-22 and the sex chromosomes are shown as pair 23.**

Karyotype of a Normal Female



Karyotype of Male with Down Syndrome

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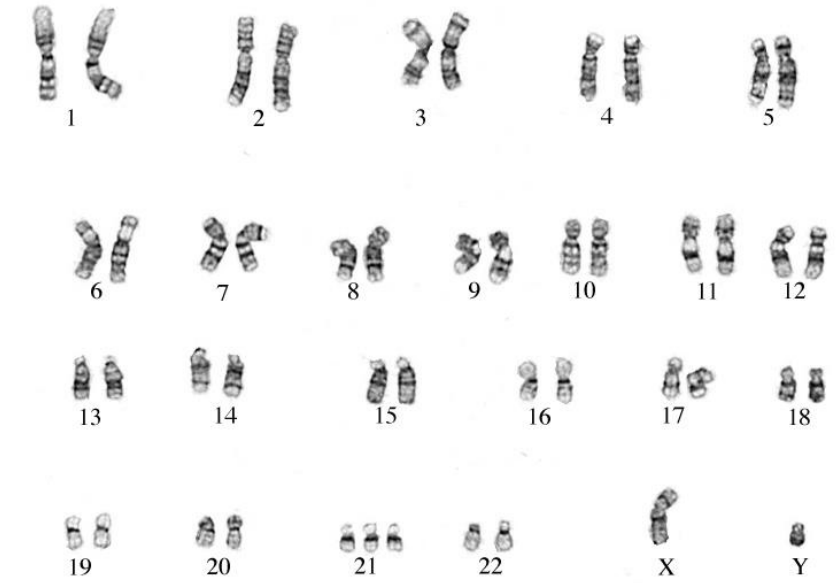
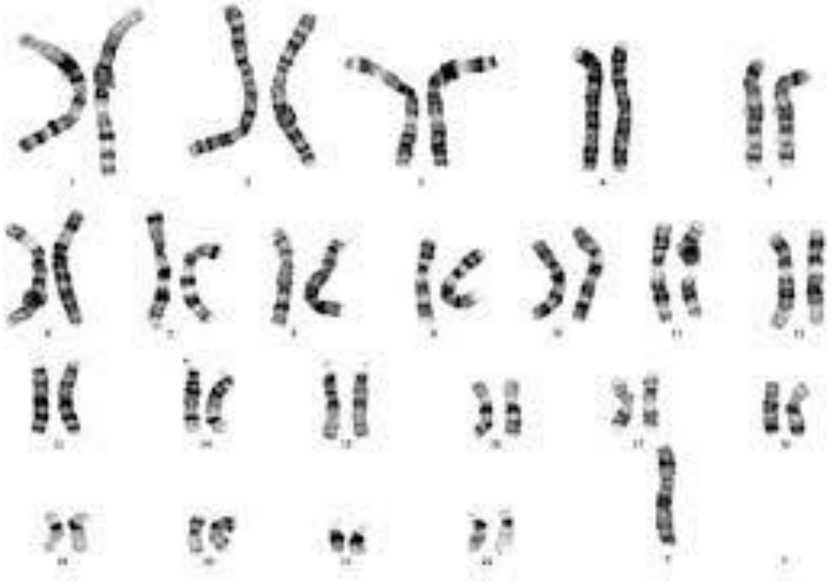


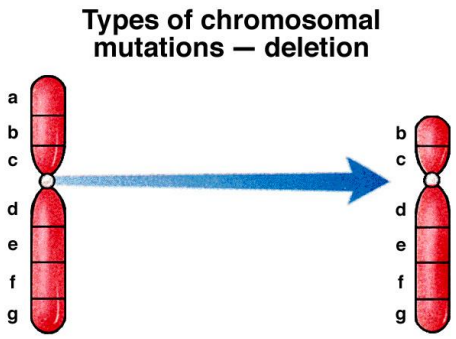
Karyotype of a Female with Turner Syndrome

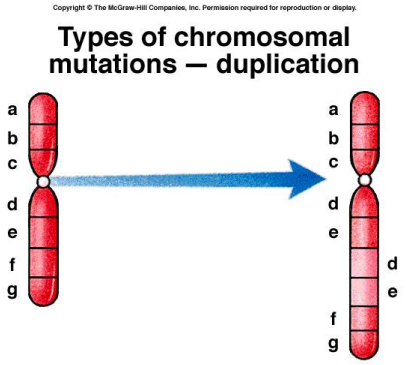


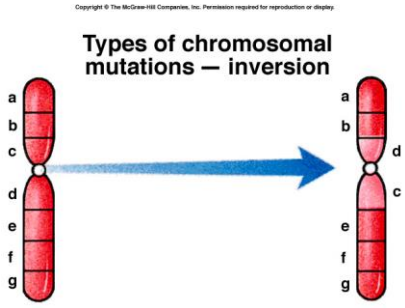
Chromosomal Rearrangements

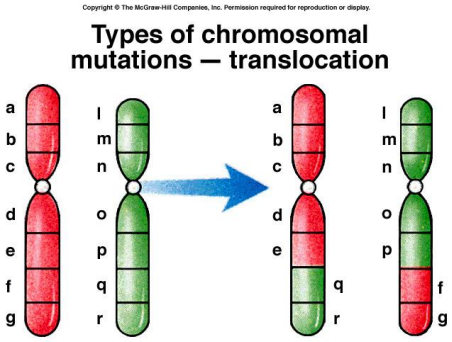
In some cases, large chunks of chromosomes (but not entire chromosomes) are affected. Such changes are called **chromosomal rearrangements or chromosomal mutations**. The rearranged chromosomes are called **aberrant chromosomes. The rearrangements often happen when crossing over doesn't work correctly. There are four main types of chromosomal rearrangements.**

Duplication—A chromosome ends up with two or more copies of a gene segment.
D**eletion—**A chromosome ends up with no copies of a particular gene segment.
I**nversion**—A chromosomal region is flipped around so that it points in the opposite, wrong direction.
Translocation-- A piece of one chromosome is attached to another non-homologous chromosome.









Chromosomal rearrangements are responsible for several human disorders. These include:

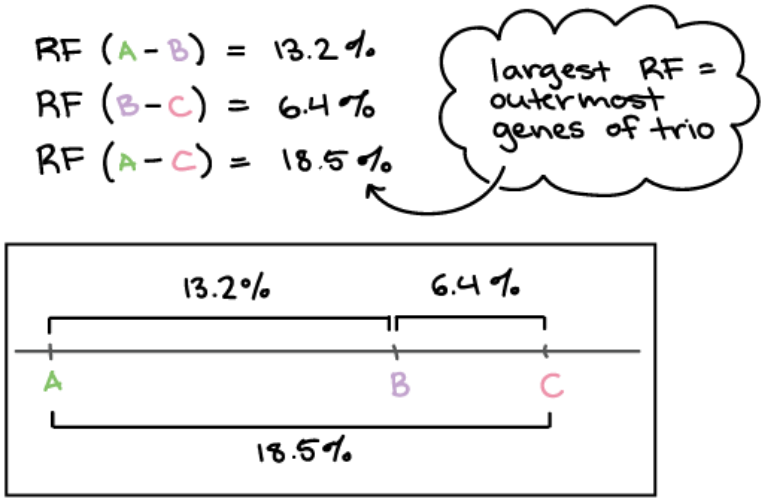
Deletions—**Cri Du Chat**—Part of chromosome 5 is deleted. This leads to symptoms which include a high-pitched cat-like cry, **mental retardation**, delayed development, distinctive **facial** features, small head size (microcephaly), widely-**spaced** eyes (hypertelorism), low birth weight and weak **muscle** tone (hypotonia) in infancy.
Duplications—Fragile X syndrome—part of the X chromosome is duplicated. This leads to mental retardation.
Inversions—may not cause any problems—can lead to infertility
Translocations—**Philadelphia chromosome—A segment of** chromosome 9 is translocated to chromosome 22. This leads to a severe form of leukemia.

Crossover frequency

Genes that are located on the same chromosome are said to be linked. If the genes are located far apart from each other, they appear to independently assort due to crossing over. If the genes are located near each other, they don't independently assort and are usually inherited together.

Scientists often use the crossover frequency of linked genes to estimate the order of the genes on the chromosome and their relative distances from each other. The crossover frequency is essentially a measure of how often two genes that start out on the same chromosome end up on opposite chromosomes after crossing over.

For example, let's suppose we have three linked genes, *A*, *B*, and *C*, and we want to know their order on the chromosome (*ABC*? *ACB*? *CAB*?) If we look at recombination frequencies among all three possible pairs of genes (*AC*, *AB*, *BC*), we can figure out which genes lie furthest apart, and which other gene lies in the middle. Specifically, the pair of genes with the largest recombination frequency must flank the third gene:



By doing this type of analysis with more and more genes (e.g., adding in genes *D*, *E*, and *F* and figuring out their relationships to *A*, *B*, and *C*) we can build up linkage maps of entire chromosomes. In linkage maps, you may see distances expressed as centimorgans or map units rather than recombination frequencies. Luckily, there's a direct relationship among these values: a 1 percent recombination frequency is equivalent to 1 **centimorgan** or 1 **map unit**.

Mitosis and Meiosis Compared and Contrasted

Mitosis and meiosis are both forms of nuclear division. They are similar in the way the chromosomes segregate but differ in the number of cells produced and the genetic contents of the resulting cells. The table below summarizes the key similarities and differences of the two processes.

Mitosis	Meiosis
1. One division completes the process	1. Two divisions are required to complete the process
2. Chromosomes do not synapse.	2. Homologous chromosomes synapse in prophase
3. Homologous chromosomes do not cross over.	3. Homologous chromosomes do cross over.
4. Centromeres divide in anaphase.	4. Centromeres divide in anaphase II, but not in anaphase I
5. Daughter cells have the same number of chromosomes as the parent cell (2n→2n)	5. Daughter cells have half the number of chormosomes as the parent cell. (2n→n)
6. Daughter cells have the same genetic information as the parent cell	6. Daughter cells are genetically different from the parent cell.
7. Results in growth, replacement of worn-out cells, and repair	7. Results in sex cells.

Mendelian Inheritance

Gregor Mendel—Mendel is considered to be the "Father of Genetics" because of his groundbreaking work with inheritance in pea plants. Mendel conducted his experiments at an Austrian monastery in the late 1800s. He discovered the basic laws of inheritance. He was able to do this before science had an understanding of DNA, chromosomes, or meiosis. One of the characteristics of Mendel's work that made it so successful was that he quantitatively analyzed the results of his pea plant breeding experiments. Most biologists, before Mendel, didn't think that biological processes could be mathematically analyzed. Mendel's main conclusions included:

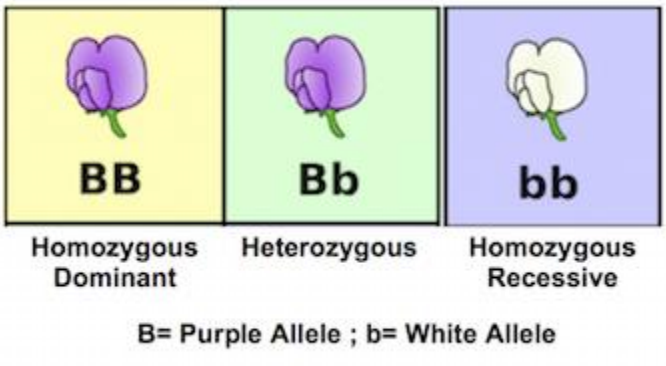
1. Each trait is controlled by a single pair of alleles. Mendel called them factors.
2. Some alleles are dominant and can mask the appearance of recessive alleles (**Law of Dominance**)
3. The alleles from a pair separate (during meiosis) and only one member of each pair is transmitted to an offspring from each parent (**Law of Segregation**). Realize that Mendel did not know about meiosis, but essentially predicted its existence.
4. The maternal and paternal alleles/factors from each pair assort independently from the alleles in the other pairs. All possible combinations of alleles/factors occur in the gametes. We know today that independent assortment occurs during Metaphase I of Meiosis. (**Law of Independent Assortment**).

We know today that Mendel's Laws of Segregation and Independent Assortment apply only to genes that are located on different chromosomes. Mendel was unaware of the existence of chromosomes and linked genes.

Important Terms

Trait—a genetically determined characteristic. Mendel hypothesized that each trait is determined by two alleles (one inherited from each parent). Although this is often true, there are many exceptions to this rule. We will explore some of those exceptions in the Non-Mendelian Inheritance section of the notes (included below).

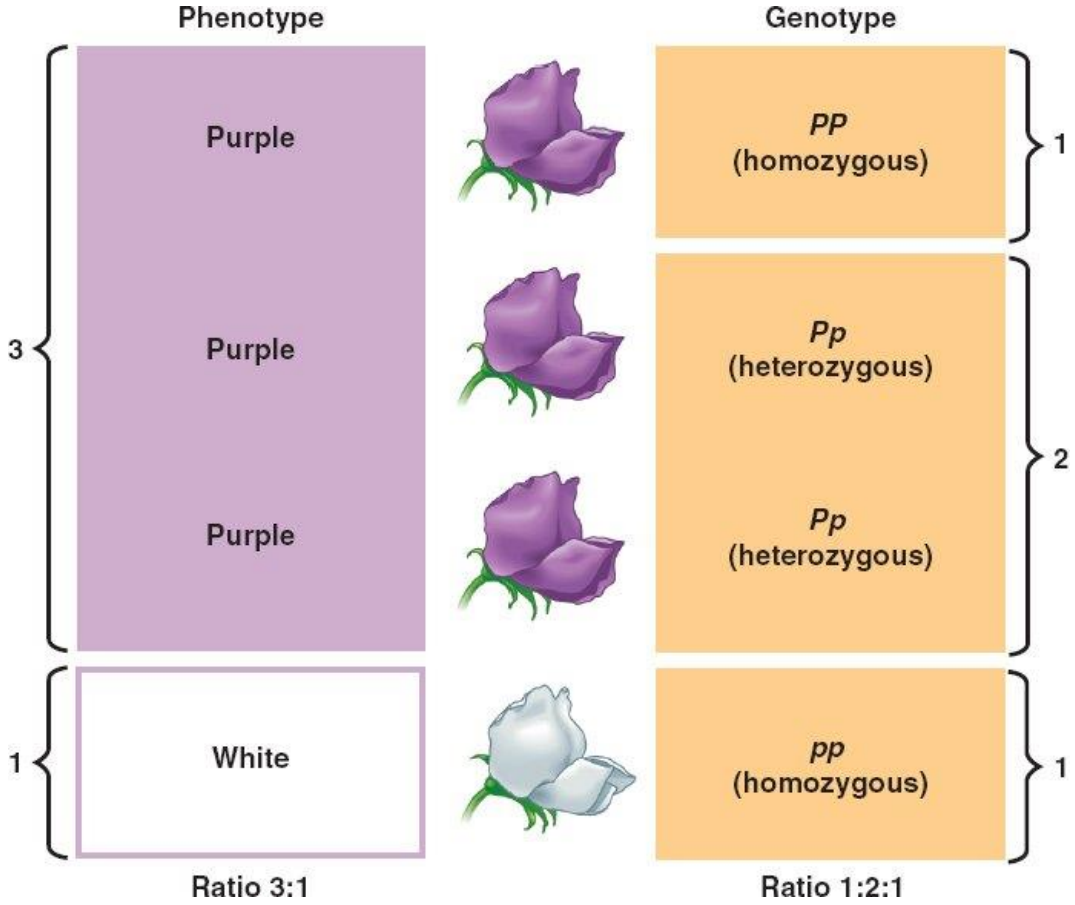
Alleles--This term refers to **different versions of a gene**. For example, a gene might control the color of one's eyes. Different versions (alleles) of that gene might cause the individual to have brown, blue, or green eyes. **Dominant** alleles are represented with capital letters, while r**ecessive** alleles are represented with lower case letters.



Homozygous—An organism is said to be homozygous or **pure bred** for a trait if the two alleles it possesses for the trait are identical. (TT or tt for example).

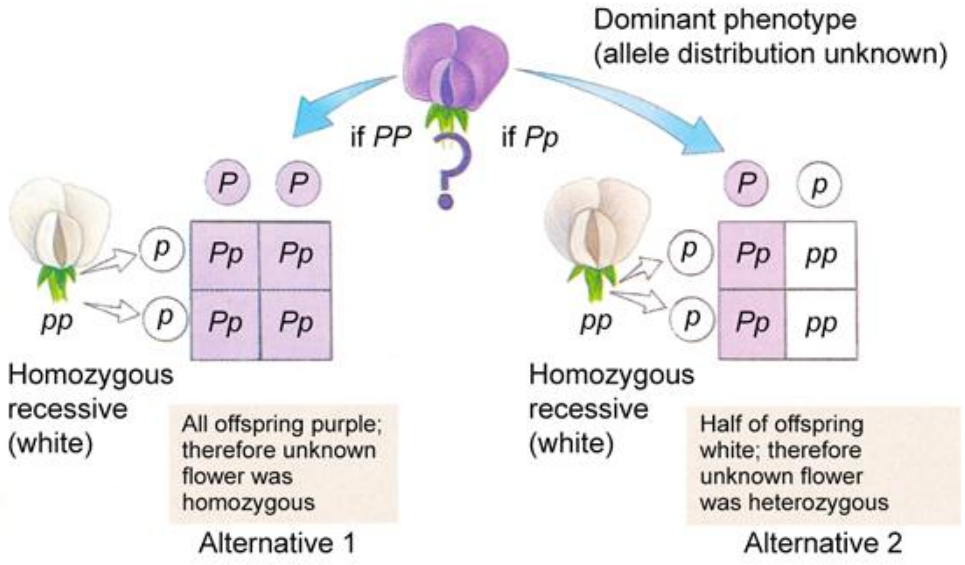
Heterozygous-- An organism is said to be heterozygous or a **hybrid** for a trait if the two alleles it possesses for the trait are different. (Tt for example).

Phenotype--This term refers to the physical traits or appearance of an organism. (Blue eyes or Type A blood, would be examples.)



Genotype--This term refers to an organism's genetic (DNA) make-up for a trait. (Such as TT, Tt or tt.) A genotype of TT might cause a pea plant to have the "tall" phenotype.

If the genotype of an organism is unknown, we can perform a **TESTCROSS** to determine it. To perform this test, you must cross the individual with the unknown genotype with a **homozygous recessive individual**. The phenotypes of the resulting offspring can then be used to deduce the genotype of the parent.



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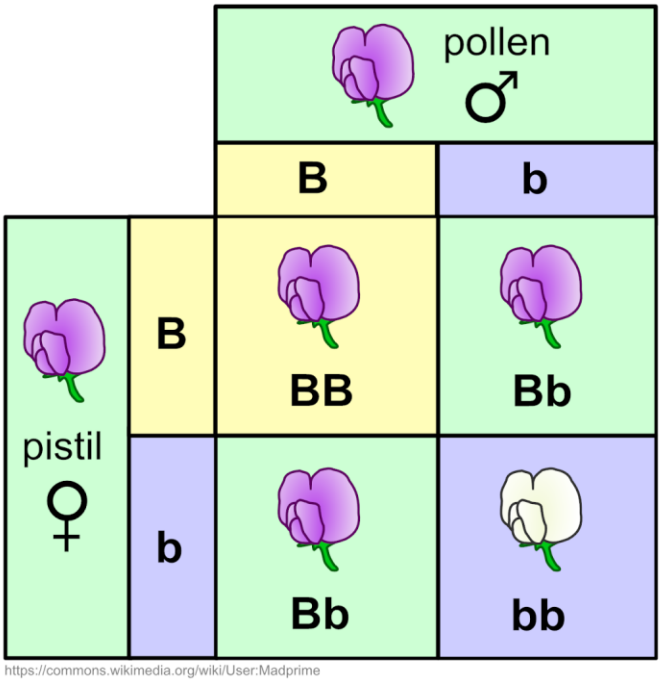
Punnett Square--This is a chart which shows the **possible** genotypic outcomes for a mating cross based on the parents' genotypes.

Monohybrid Cross

Monohybrid Cross—This is a cross in which only one trait is analyzed. To complete the cross, you must first determine the possible gamete combinations of the parents. Remember that gametes are haploid. They should only contain one allele for each trait. For example: If we crossed two purple flowered plants, with the genotype Bb, the possible gametes for each plant are "B" or "b". The gametes for one parent are written along the top of the Punnett square, one haploid gamete per column. The gametes of the other parent are written along the left hand side of the Punnett square, one haploid gamete per row. The boxes inside the square are then filled in using the column and row headers as shown below.



https://commons.wikimedia.org/wiki/User:Madprime

The cross illustrated above predicts a **phenotypic ratio** of 3 purple: 1 white and a **genotypic ratio** of 1 BB: 2Bb:1 bb

The rules of probability can be applied to analyze the passage of single-gene traits from parent to offspring. The two rules commonly used in an AP Biology class are listed below:

If A and B are mutually exclusive (separate, unconnected events) then:

P(A or B)=P(A) + P (B)
The rule listed above is used to calculate the probability (P) of either event A or event B happening. The probability that either event will occur is simply the sum of each of the individual events.

Product Rule

P (A and B)=P(A) X P(B)
The product rule is used to calculate the probability (P) that multiple independent events will each occur. The probability that both event A and event B will occur is the product of the individual probabilities.

If you were asked to determine the **probability** that the cross above would produce a plant with white flowers, your answer would be ¼ or 25% (from the Punnett square). If you were asked to determine the probability that the cross would produce a plant with purple flowers, your answer would be ¾ or 75%.

If you were asked to determine the probability that the above cross would produce **either** a homozygous purple (PP) or a homozygous white (pp) plant, you would: First use the Punnett Square to determine that the probability of getting a homozygous purple plant is ¼ and the probability of getting a homozygous white plant is also ¼. Then use:
P(A or B)= ¼ + ¼= ½

Suppose you were asked to determine the probability that the cross produced two purple-flowered plants in a row?
To calculate this probability, you would need to use the **product rule**. The product rule states that in order to determine the probability of two independent events occurring together, you must multiply the individual probabilities of each event. In this example, we should multiple ¾ X ¾ to get a probability of 9/16 or 56.25%.

The Product Rule

- If two or more events are independent of one another, the likelihood of their simultaneous or consecutive occurrence is the product of their individual probabilities
- This is the **product rule,** also called the **multiplication rule**

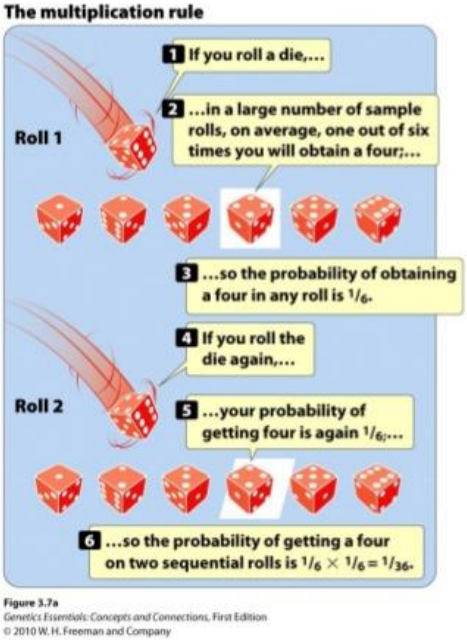


Figure 3.7a
Genetics Essentials: Concepts and Connections, First Edition
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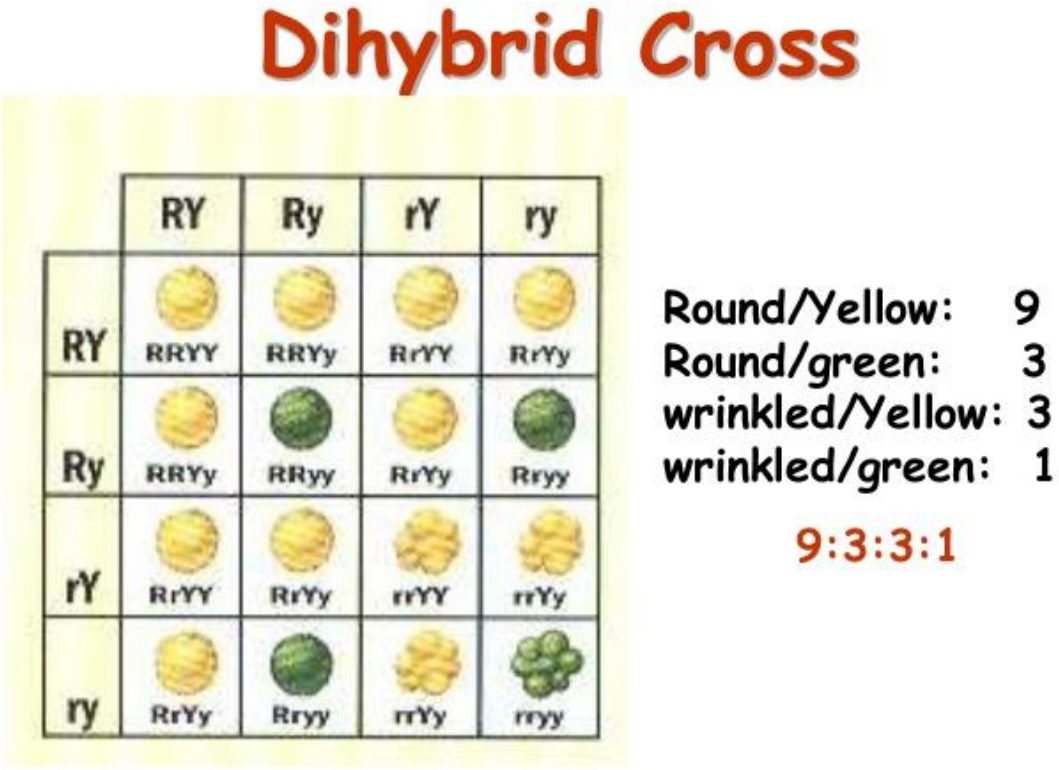
Dihybrid Cross

Dihybrid Cross–A dihybrid cross is a cross in which **the inheritance of two traits is analyzed at the same time.** For example: Suppose that in pea plants "R" is the allele for round seeds, "r" is the allele for wrinkled seeds, "Y" is the allele for yellow seeds, and "y" is the allele for green seeds. The Punnett Square below illustrates the cross between two plants that are heterozygous (RrYy X RrYy) for both traits, **a true dihybrid cross**. To construct the cross, you must first determine the possible gametes that each parent can create**. Each gamete should contain only one allele from each gene.** In this case, each allele contains only one R (either upper or lowercase) and one Y (either upper or lowercase). All possible combinations of the parental alleles are possible. This means that each of the two parents can create the following gamete combinations: RY; Ry; rY; and ry.

To determine the possible number of unique gamete combinations, analyze each gene pair. For the R pair, each parent has a single R and a single r, **2 different alleles.** For the Y pair, each parent also has a single Y and a single y**, 2 different alleles.** To calculate the number of possible unique gametes, multiply the number of unique alleles in each gene pair together. In this case that would mean 2 (from the R pair) X 2 (from the Y pair) =**4 unique gametes per parent.**

Let's say that one of the original parents had the following genotype: **rrYy** To calculate the number of possible alleles from this parent, multiple 1 (from the unique alleles in the r pair) X 2 (from the unique alleles in the Y pair) to get **2 possible unique gamete combinations**. In this case those would be **rY and ry.**

Once the possible gamete combinations are determined, the gametes for one parent are written along the top of the Punnett square, one haploid gamete per column. The gametes of the other parent are written along the left-hand side of the Punnett square, one haploid gamete per row. The boxes inside the square are then filled in using the column and row headers as shown below.



In a true dihybrid cross (one in which both parents are heterozygous for both traits), the phenotypic ratio will always be 9:3:3:1 as shown in the Punnett square above.

Trihybrid or Larger Cross

It is possible to construct Punnett Squares for trihybrid crosses (crosses in which the inheritance of three traits are analyzed at the same time). These squares can be really large (up to 64 squares). You will typically not be asked to draw a trihybrid Punnett Square on the AP exam. You might, however, be asked to answer questions like the following:

How many different possible gametes can an individual with the following genotype produce Aa Bb Dd Ee?

To answer this question, use the same approach as that described above. Multiple the number of unique alleles from each pair together. In this case. 2 unique alleles from the A pair X 2 unique alleles from the B pair X 2 unique alleles from the D pair X 2 unique alleles from the E pair to yield 16 total unique allele combinations/gamete types.

If the genotype of the parent had instead been AA Bb cc, the calculation would have been:
1 unique allele from the A pair X 2 unique alleles from the B pair X 1 unique allele from the c pair to yield only two possible gamete combinations. In this case, those combinations would be ABc and Abc.

What is the probability that the cross between the following genotypes:
Aa BB Dd Ee X Aa Bb Dd ee will produce an offspring with the genotype Aa BB dd Ee?

To answer this question, you could draw a huge Punnett Square, but the easiest and fastest approach is to use the product rule. First, analyze each gene pair separately. Think of doing four separate Punnett squares.

What is the probability that AA (from parent 1) and Aa (from parent 2) will yield (Aa) in the offspring? **The answer is ½.**

What is the probability that BB (from parent 1) and Bb (from parent 2) will yield BB in the offspring? Again, **the answer is ½.**

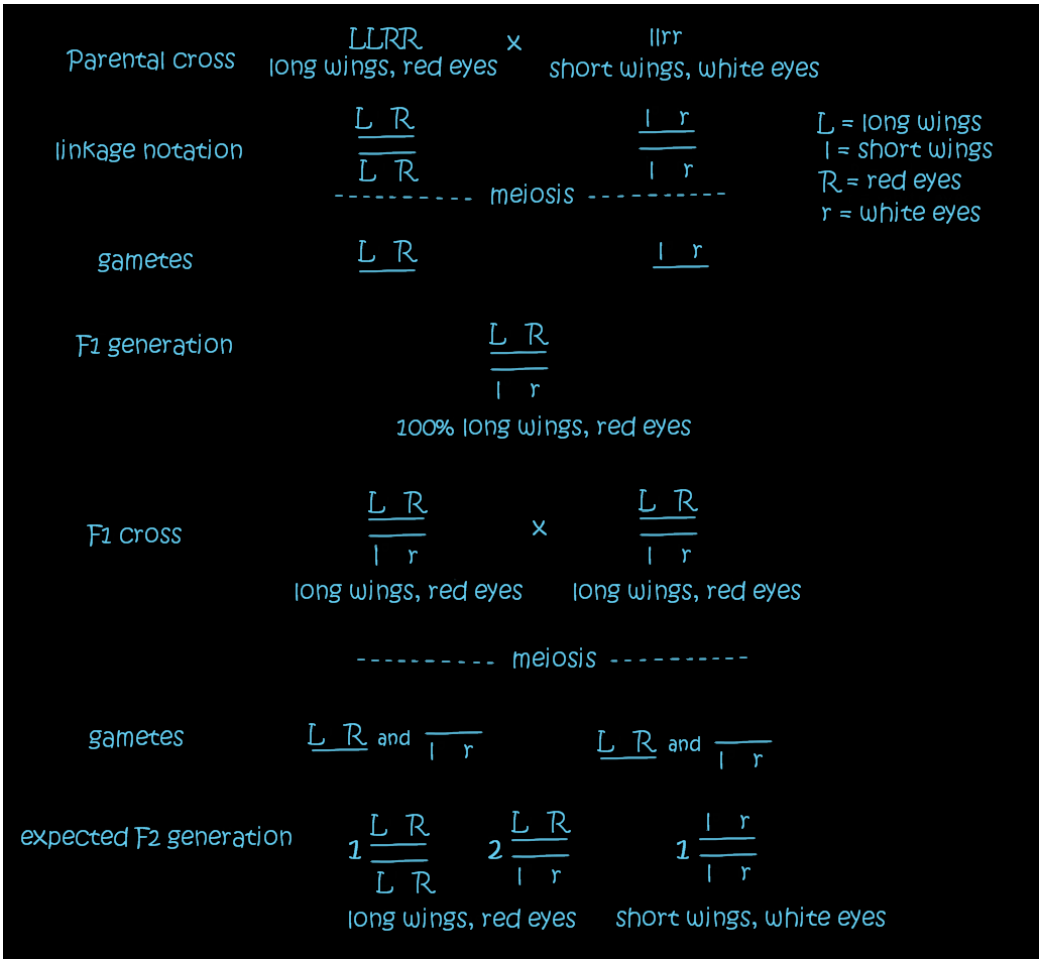
What is the probability that Dd (from parent 1) and Dd (from parent 2) will yield dd in the offspring? This time, **the answer is ¼.**

What is the probability that Ee (from parent 1) and ee (from parent 2) will yield Ee in the offspring? This time **the answer is ½.**

To determine the overall probability that the cross between parents Aa BB Dd Ee X Aa Bb Dd ee will produce an offspring with the genotype Aa BB dd Ee, multiply the probabilities from each gene pair (in bold above). In this case, the calculation is ½ (from the A pair) X ½ (from the B pair) X ¼ (from the C pair) X ½ (from the D pair) **to yield an overall probability of 1/32.**

Linked Genes and Dihybrid Crosses

Linked genes occur on the same chromosome, and therefore, tend to be inherited together (i.e., do not segregate independently). When two heterozygotes are mated in a normal dihybrid cross with independent assortment of alleles, the expected ratio in the offspring is 9:3:3:1. However, as shown in the figure below, in cases of dihybrid crosses involving linkage, the ratio of the offspring produced is 3:1 and only the parental types, with no recombinants, are expected.



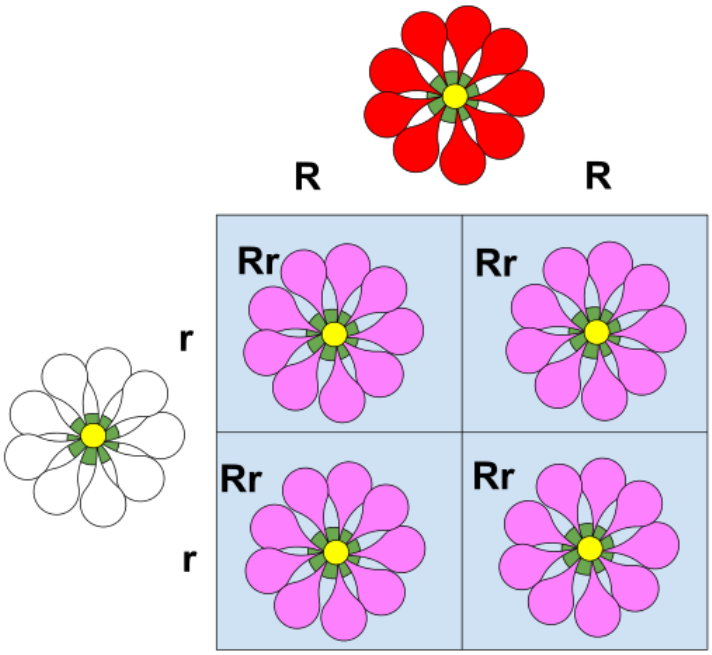
The table below includes the actual observed results of the dihybrid cross involving the two heterozygotes described above. Even though offspring with long wings/white eyes and offspring with short wings/red eyes weren't expected, some were produced. **This often occurs in crosses involving linked genes because of the genetic recombination that occurs during crossing over.**

observed F2 generation	$\frac{L\ R}{l\ r}$	$\frac{L\ r}{l\ r}$	$\frac{l\ R}{l\ r}$	$\frac{l\ r}{l\ r}$
	12	2	4	13
	long wings, red eyes	long wings, white eyes	short wings, red eyes	short wings, white eyes

Non-Mendelian Inheritance

Although Mendel discovered some of the most basic laws of inheritance, scientists have discovered many exceptions to the so-called laws of Mendelian inheritance. The processes/conditions described below are all examples on **non-mendelian inheritance patterns. In these situations, the patterns of inheritance do not follow the ratios predicted by Mendel's laws. Quantitative analysis of the results of such crosses reveals that the observed phenotypic ratios statistically differ from the ratios predicted by Mendel's laws.**

Incomplete Dominance is a form of intermediate inheritance in which one allele for a specific trait is not completely expressed over its paired allele. This results in a third phenotype in which the expressed physical trait is a combination or **blending** of the phenotypes of both alleles. In some plants, there are alleles for red flowers (R) and for white flowers (r). Heterozygous plants (Rr) end up with pink flowers (a blend between red and white).

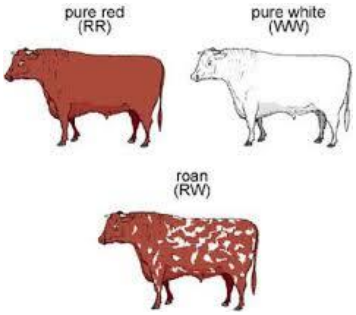


Codominance—This is a condition in which both alleles in a pair are expressed at the same time. They are both equally present in the phenotype (not blended). An example of codominance is human blood type. There are alleles for Type A blood (I^A), Type B (I^B), and type O (i) blood. The alleles for type A and B code for different cell surface proteins that occur on the surface of the red blood cells (RBCs). Individuals with two O alleles (ii) lack these proteins. Individuals with either I^AI^A or I^Ai possess the type A proteins on their RBCs. Individuals with either I^BI^B or I^Bi possess the type B proteins on their RBCs. Individuals with the I^AI^B genotype possess both the A and B proteins on the surface of their RBCs.

Genotype	Red blood cell appearance	Phenotype (blood group)
I^AI^A or $I^A\,i$		A
I^BI^B or $I^B\,i$		B
I^AI^B		AB
ii		O

Another commonly cited example of codominance is the inheritance pattern of coat coloration in some cows.

There are two unique alleles (R) for red coat color and (W) for white color. Cows that inherit one of each allele (RW) have a blotchy phenotype which includes both red and white patches. This type of coloration is known as **roan**.



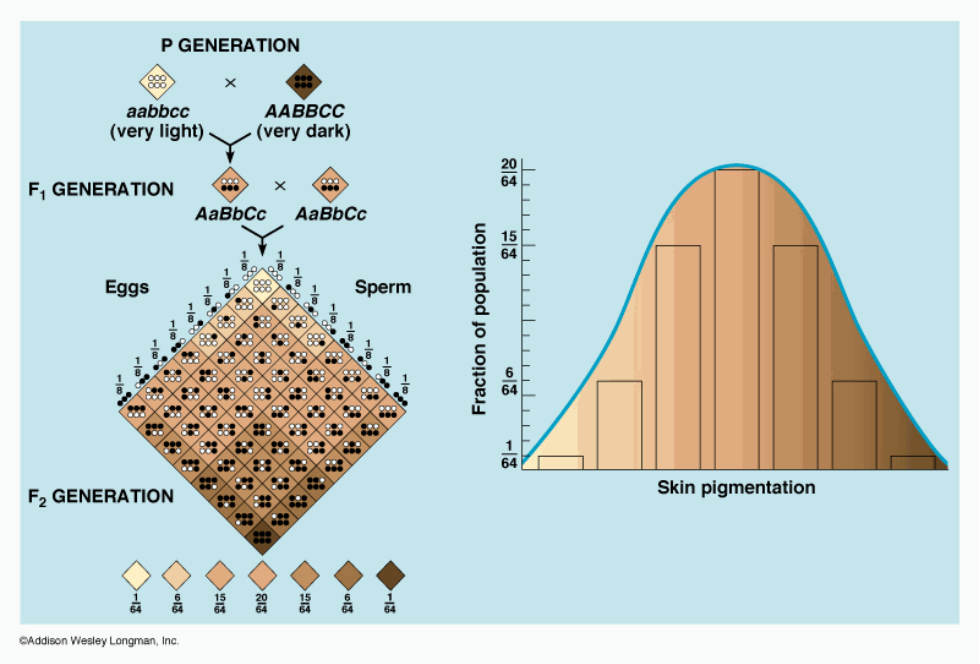
Multiple Alleles—This is when three or more alternative forms of a gene (**alleles**) can occupy the same **locus (location)**. However, only two of the **alleles** can be present in a single organism. For example, the ABO system of blood types is controlled by three **alleles (I^A, I^B, i)**, only two of which are present in an individual.

Pleiotropy is a condition in which one gene affects multiple (seemingly unrelated) characteristics.

> **Sickle Cell Disease** is a great example. The gene AFFECTS the shape of one of the polypeptide chains that make up **hemoglobin** (the molecule that allows red blood cells to transport oxygen). The change in the polypeptide's shape also causes the red blood cells to change shape (from round to sickle shaped). This impedes the flow of blood and leads to multiple symptoms all over the body such as anemia, physical weakness, impaired mental function, lowered disease resistance, and paralysis**. Individuals who are heterozygous for the sickle cell mutation also possess resistance to the protozoan that causes malaria.** This **heterozygote advantage** explains why the normally harmful mutation is so common in human populations where malaria has consistently been a problem.

Epistasis is a condition in which a gene at **one locus** affects a gene at a **second locus.** Albinism in humans is a good example of epistasis. Albinism arises when individuals inherit two defective copies of the allele that normally codes for the enzyme necessary for melanin (pigment) synthesis. Even though alleles at other loci may code for brown eyes, skin, or hair, these individuals are albinos because they lack the ability to produce the pigment (melanin).

Polygenic Inheritance occurs when a trait is governed by two or more sets of alleles. Examples of human traits that are polygenic include height, skin color, and the prevalence of diabetes. Each individual possesses a copy of all the allelic pairs. These may be located on different chromosomes. Each dominant allele has a quantitative effect on the phenotype and the effects are additive. The population typically exhibits **continuous phenotypic variations**. If the frequency of the different phenotypes were graphed, the graph would look like a bell curve. Human skin color is thought to be controlled by 3 pairs of alleles (A, B, C). A person with the alleles *AABBCC* is very dark skinned, a person with *aabbcc* alleles is very light skinned, and a person with *AaBbCc* (or any combo) has an intermediate skin color.



Multifactorial traits are those controlled by multiple genes that are also affected by physiological and environmental influences. Hypertension, diabetes, schizophrenia, and allergic conditions are probably all multifactorial traits. Such traits do not segregate in the predicted Mendelian patterns.

Lethal alleles—There are certain allele combinations which are lethal and prevent the birth of individuals with certain genotypes. For example, **Achondroplasia** is an autosomal dominant form of human dwarfism. Individuals with the genotype Aa are dwarfs. Individuals with the genotype aa are normal. The genotype AA is lethal. Individuals with this genotype die before birth. The Punnett square for the cross between two dwarfs (Aa) would lead one to believe that ¾ of their children should be dwarfs.

	A	a
A	AA	Aa
a	Aa	aa

Due to the lethality of the AA genotype. the actual probability that a living dwarf child will be born is 2/3.

Pedigree Charts/Modes of Inheritance

Important Terms

Pedigree Chart--A diagram showing the lineage or genealogy of an individual and all the direct ancestors, usually to analyze or follow the inheritance of trait. The pattern of the inheritance of monohybrid, dihybrid, sex-linked, and genetically-linked genes traits can often be predicted from data, including pedigrees, which indicate the genotypes and/or phenotypes of parent and offspring.

Mode of Inheritance--The manner in which a genetic trait or disorder is passed from one generation to the next. Autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked, and mitochondrial **inheritance** are examples.

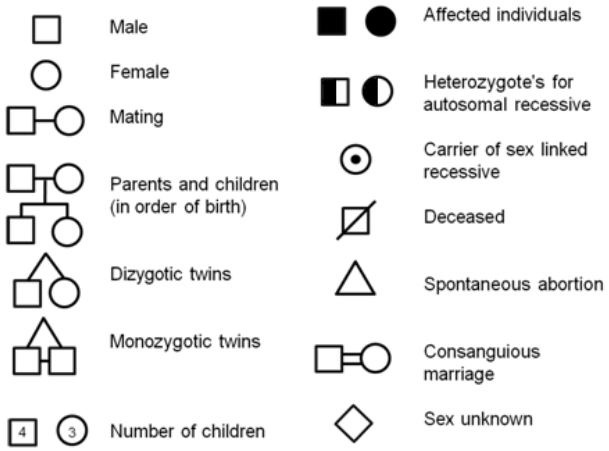
Autosome—A non-sex chromosome. Humans have 44 autosomes per diploid, somatic cell.

X and Y chromosomes—The two types of sex chromosomes in mammals, flies, and most other animals. These chromosomes determine the sex of an individual. Females usually have 2 X chromosomes per somatic cell, while males usually have only 1 X chromosome per cell. In certain species, the chromosomal basis of sex determination is not based on X and Y chromosomes. Birds, butterflies, and snakes take everything we know about sex at the chromosomal level and stand it on its head. Instead of X and Y chromosomes, they have Z and W chromosomes.What's the difference? Everything. The XY and ZW chromosomes share no genes at all. What's more, the ZW chromosomes flip the sex determination system. A male chicken - or peacock, or giant river prawn, or komodo dragon - has ZZ chromosomes. A female has ZW chromosomes. For both XY and ZW systems, there are all kinds of variations on what happens if one gene is missing or doubled. In mammals, an XO (an X chromosome and a missing sex chromosome) usually results in a female. When it comes to fruit flies or crickets, a single X chromosome results in a male. So sometimes the presence of a Y is required to produce a male, and sometimes it isn't. There haven't been any ZO birds found, indicating they need at least two chromosomes to develop, but ZO, ZZW and ZZWW all produce female butterflies. Snakes added to the confusion recently, when a female python produced offspring despite not having been near any males. This parthenogenesis produced all-female offspring with WW chromosomes — a combination not previously thought possible.

Carrier—An Individual who possesses only one copy of a recessive allele. This individual doesn't express the trait, but can pass the allele on to his/her offspring.

Gene Linkage—Genes are linked if they are found together on the same chromosome.

The diagram included below illustrates the symbols commonly used in pedigree charts.

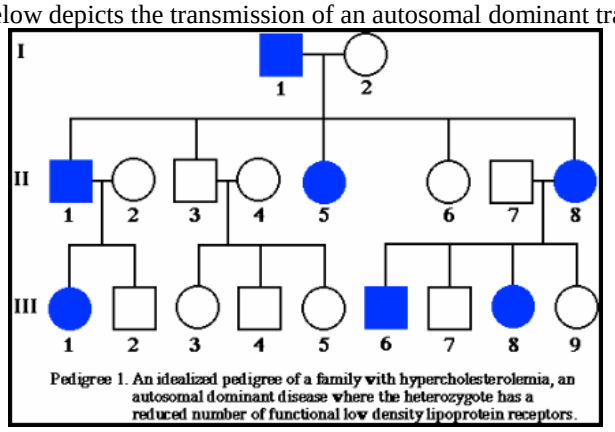


Modes of Inheritance—Autosomal Dominant

The genes for autosomal dominant traits are located on one of the 44 autosomes in a human cell. The genes are dominant, which indicates that individuals with only one copy of the gene are affected. Characteristics of autosomal dominant traits include:

- Males and females are equally likely to have the trait.
- Traits do not skip generations because there are no carriers for the traits.
- The trait is present whenever a single copy of the corresponding allele is present.
- There is male-to-male transmission. This means that fathers can pass to trait to their sons.

Some examples of human genetic conditions that are transmitted via the autosomal dominant mode of inheritance include: Huntington's Disease, Achondroplasia, Polycystic Kidney Disease, and Familial Hypercholesterolemia. The pedigree chart included below depicts the transmission of an autosomal dominant trait.



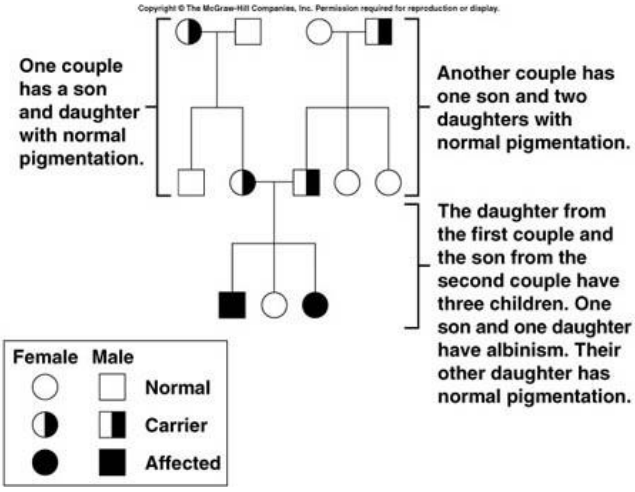
Pedigree 1. An idealized pedigree of a family with hypercholesterolemia, an autosomal dominant disease where the heterozygote has a reduced number of functional low density lipoprotein receptors.

Modes of Inheritance—Autosomal Recessive

The genes for autosomal recessive traits are located on one of the 44 autosomes in a human cell. The genes are recessive, which indicates that individuals must possess two copies of the recessive allele in order to exhibit the trait. Characteristics of autosomal recessive traits include:

- Males and females are equally likely to have the trait.
- Traits often skip generations (due to the possibility of carriers).
- Only homozygous recessive individuals have the trait.
- Traits may appear in siblings without appearing in their parents.
- If a parent has the trait, those offspring who do not have it are heterozygous carriers of the trait.
- Some examples of human genetic conditions that are transmitted via the autosomal recessive mode of inheritance include: Tay Sachs Disease, PKU, and Cystic Fibrosis.

The pedigree chart included below depicts the transmission of an autosomal recessive trait.



Modes of Inheritance—X-linked Dominant

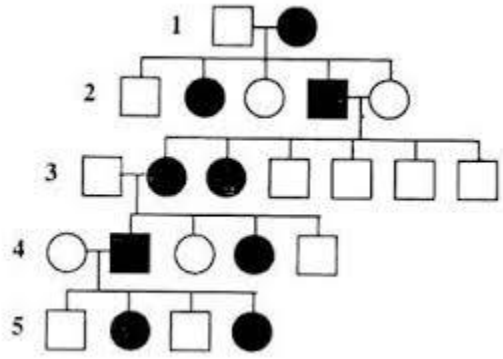
Some traits are determined by genes located on sex chromosomes (either X or Y). Such traits are known as sex-linked traits. The genes for X-linked dominant traits(a type of sex-linked trait) are located on the X chromosome in a human cell. The alleles are dominant, so individuals with only one copy of the allele exhibit the trait. **X-linked traits (whether dominant or recessive) are always expressed in males (if the male's X carries the allele) because males possess only one X chromosome.**

Characteristics of X-linked dominant traits include:

- The trait is present whenever the corresponding gene is present.
- There is no male-to-male transmission. Fathers cannot pass the trait to their sons.
- A female who has the trait may or may not pass the allele for the trait to her son or daughter.
- All of the daughters of a male with the trait will inherit the trait.

Some examples of human genetic conditions that are transmitted via the X-linked dominant mode of inheritance include: hypertrichosis, porphyria, and Rett syndrome.

The pedigree chart included below depicts the transmission of an X-linked dominant trait.

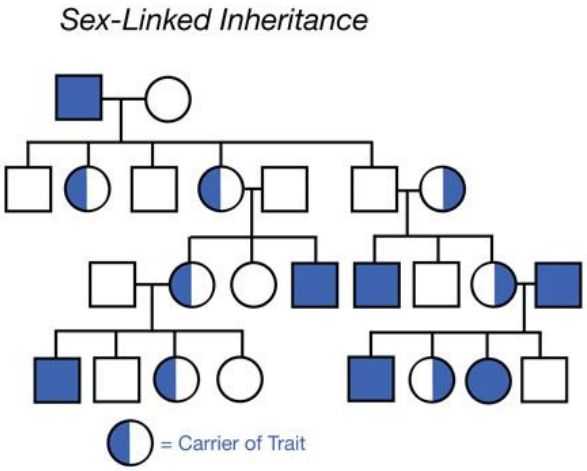


Modes of Inheritance—X-linked Recessive

The alleles for X-linked recessive traits are located on the X chromosome in a human cell. The alleles are recessive, so for a female to exhibit the trait she must possess the allele on both of her X chromosomes. Since a male has only one X chromosome, he is affected if his X chromosome carries the allele. Characteristics of X-linked recessive traits include:

- These traits are far more common in males than in females.
- Traits may skip generations.
- All daughters of a male who has the trait are either affected or are heterozygous carriers.
- The son of a female carrier has a 50 percent chance of having the trait.
- Mothers of males who have the trait are either heterozygous carriers or homozygous and express the trait.
- There is no male-to-male transmission. Fathers cannot pass the trait to their children.
-

Some examples of human genetic conditions that are transmitted via the X-linked recessive mode of inheritance include: hemophilia, red/green colorblindness, Duchenne Muscular Dystrophy, and Lesch-Nyhan syndrome.
The pedigree chart included below depicts the transmission of an X-linked recessive trait.



Inheritance of Red-Green Blindness:
an X-linked Recessive Trait

Modes of Inheritance—Y-linked

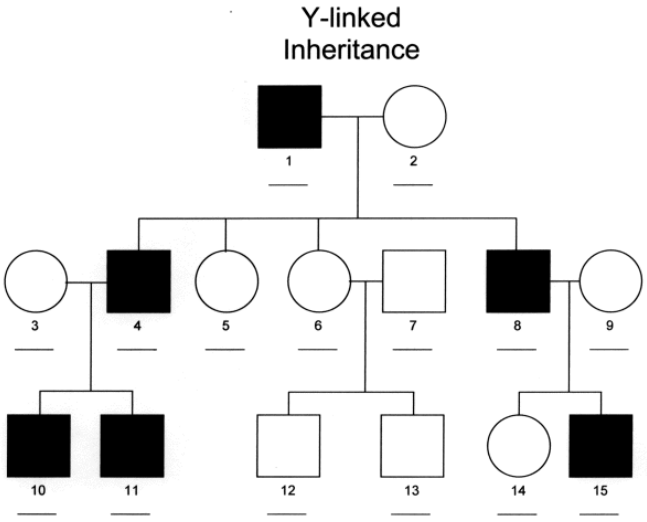
The alleles for Y-linked traits are located on the Y chromosome in a human cell. Since males possess only one Y chromosome, the traits aren't usually referred to as dominant or recessive. A man who possesses a Y-linked allele will exhibit the trait it controls. Since females don't possess a Y chromosome, they are not affected by Y-linked traits.

Characteristics of Y-linked traits include:

- Only males are affected.
- All sons of an affected male will also be affected.
- Females can't possess or pass on the trait.

Some forms of Retinitis pigmentosum (a form of progressive blindness) are transmitted via the Y-linked mode of inheritance

The pedigree chart included below depicts the transmission of an Y-linked trait.



Modes of Inheritance—Mitochondrial

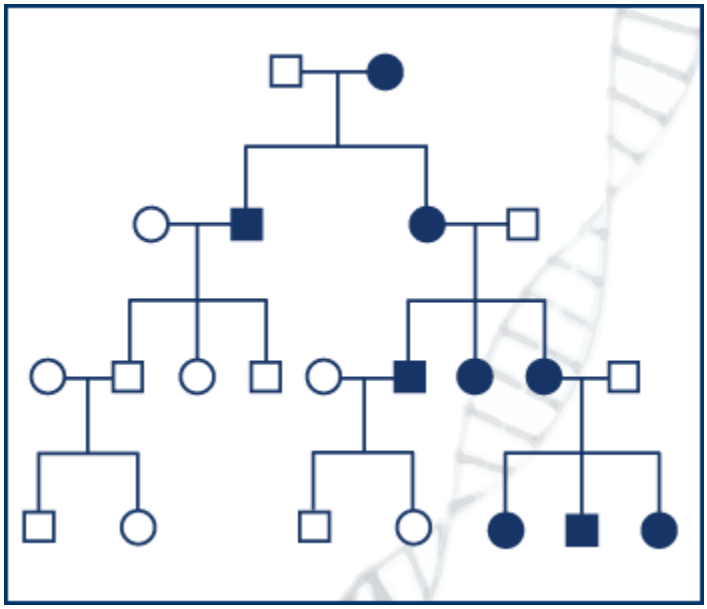
Some traits result from non-nuclear inheritance. The alleles for mitochondrial traits are located on the small circular chromosome found in the mitochondria. Since children (in animals) of both sexes inherit their organelles (including the mitochondria) from the egg of the mother, mitochondrial traits are always transmitted from mother to child. Since the mitochondria only have one chromosome each, there are no dominant or recessive mitochondrial traits. Individuals who possess a single copy of a mitochondrial allele express the trait. Characteristics of mitochondrial traits include:

- All of the children of an affected female will express the trait. These traits are always maternally inherited.
- Males can express the trait but are unable to pass it on to their offspring.

The human mitochondrial chromosome is a very small piece of DNA and codes for only 13 proteins which are part of the electron transport chain. Disorders related to mutations in the mitochondrial chromosome usually affect one's ability to make ATP.

In plants, both mitochondria and chloroplasts are transmitted in the ovule and not in the pollen. Since both organelles contain their own single, circular chromosome, traits determined by the genes on the mitochondrial and chloroplast chromosomes are always maternally inherited. Because chloroplasts and mitochondria are randomly assorted to gametes and daughter cells, traits determined by chloroplast and mitochondrial DNA do not follow simple Mendelian rules.

The pedigree chart included below depicts the transmission of a mitochondrial trait.



Environmental Effects on Phenotypes

Environmental factors influence gene expression and can lead to **phenotypic plasticity**. Phenotypic plasticity occurs when individuals with the same genotype exhibit different phenotypes in different environments. Natural environmental influences include the phenomenon of color change in the Arctic fox (and other arctic animals) from red-brown in the summer months to pure white during the winter season for better camouflage. The genes that produce the red-brown summer pigment are blocked by cold temperatures, causing the hair to grow with no color (therefore, white). Another colorful example is the interaction between the color of the hydrangea flower, which is blue in acidic soils and pink in alkaline soils. A recent study also linked improved diet in infants and adolescents to a taller average height in the United States and Europe with the opposite effect in famine-stricken populations. Some reptiles such as crocodilians and some turtles are known to display temperature-dependent sex determination (TSD), where the ambient temperature of the developing eggs determines the individual's sex. For example, in the American alligator's eggs, incubation at 33 ºC produces mostly males, while incubation at 30 ºC produces mostly females. The genetic complement of an individual is inherited; however, the environmental effect on these genes may alter their application and expression.