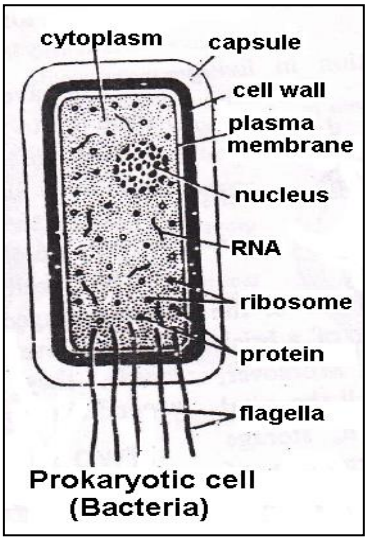
Prokaryotic Cell:

Characteristics of Prokaryotic Cells: It consists of following characteristics:

1. Prokaryotic cell has a single membrane system
2. Cell wall surrounds the plasma membrane is made up of peptidoglycan and murein
3. Cell membrane bears respiratory enzymes.
4. Cell membrane may infold to form mesosomes
5. Cell membrane helps in distributing replication products of the chromosome between the daughter cells.
6. Cytoplasm lacks membrane bound organelles like mitochondria, lysosomes, endoplasmic reticulum, golgibodies etc.
7. Ribosomes are 70s lie free in the cytoplasm or joined to the membrane
8. Cell cycle is short in prokaryotic cell taking about 20 to 60 minutes to complete.
9. Nuclear material is not enclosed by nuclear envelope and lies directly in the cytoplasm. It is called nucleoid. It contains a single prochromosome.
10. Most prokaryotes reproduce asexually through a process called binary fission. During binary fission, the single DNA molecule replicates and the original cell is divided into two identical cells.
11. There is no streaming movement occur in the cytoplasm.
12. Transcription and translation occurs in the cytoplasm.
13. Prokaryotic cells are haploid.
14. Nucleolus is absent in prokaryotic cell.

Structure of Prokaryotic Cell: It has the following structures

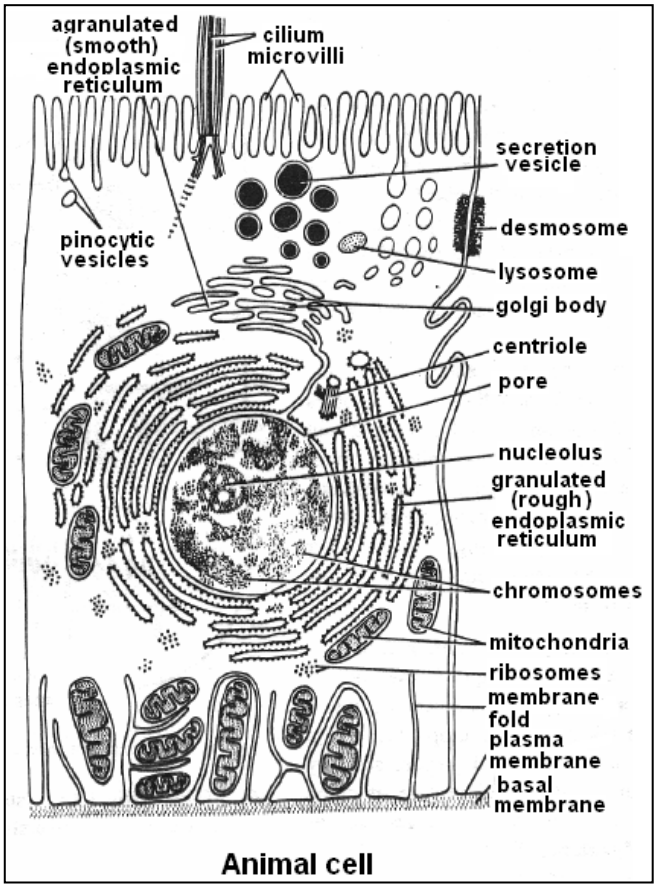


- **Capsule:** Found in some bacterial cells, this additional outer covering protects the cell when it is engulfed by other organisms, assists in retaining moisture, and helps the cell adhere to surfaces and nutrients.
- **Cell Wall**: it is the utter covering that protects the bacterial cell and gives it shape and is made up of peptidoglycan and murein.
- **Cytoplasm:** A gel-like substance composed mainly of water also contains enzymes, salts, cell components, & some organic molecules.
- **Cell Membrane or Plasma Membrane**: Surrounds the cell's cytoplasm and regulates the flow of substances in and out of the cell.
- **Ribosomes:** Cell structures responsible for protein production. It is 70s type.
- **Plasmids**: Gene carrying, circular DNA structures are not involved in reproduction.
- **Nucleoid Region**: Area of the cytoplasm contains a single bacterial DNA molecule.
- **Pili**: Hair-like structures on the surface of the cell that attach to other bacterial cells. Shorter pili called fimbriae help bacteria attach to surfaces.
- **Flagella:** Long, whip-like protrusion that aids in cellular locomotion

Eukaryotic cells:

The eukaryotic cells are the true cells which occur in plants and the animals (from Protozoa to mammals). All the cells typically composed of plasma membrane, cytoplasm and its organelles, i.e., mitochondria, endoplasmic reticulum, ribosomes, Golgi complex, etc, and a true nucleus. Here the nuclear contents such as DNA, RNA and nucleoproteins remains concentrated and separated from the cytoplasm by the thin perforated nuclear membrane.

Shape of Cells: The animal cell is spherical in shape but the shape of the cell may be irregular, triangular, tubular, cuboidal, polygonal, cylindrical, oval, rounded or elongated in different animals and plants. The shape of the cells may vary from animal to animal and from organ to organ. Cells of the same organ may display various shape.



Animal cell

Size of Cells: Mostly the eukaryotic cells are microscopic in size, but they are larger in size than the bacterial cells. The size of cells varies from 1µ to 1,75,000µ. The ostrich egg cell is usually considered as largest cell (175 mm in diameter). But certain largest cells in nerve cells have been found to have the length of 92cm to 1.06 meter.

Number of Cells: The unicellular or acellular animals (protozoan) consist of single cell. Most of the animals and many cells in the body and are known as multicellular animals or plants.

The number of cells in the multicellular organisms usually remain correlated with size of the organisms. Therefore small-sized organisms have less number of cells in comparison to large sized organisms

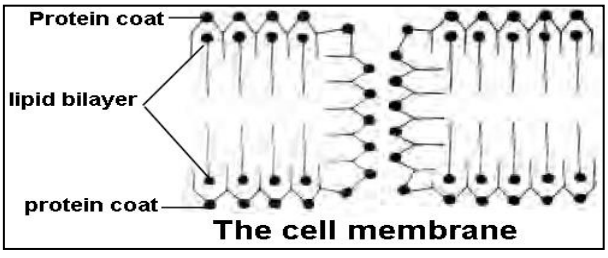
The cell consists of the following parts:

1. Plasma membrane
2. Cytoplasm; and
3. Nucleus.

Plasma Membrane: The plasma membrane is a living, ultra-thin, elastic, porous and semi-permeable membranous covering of the cell. The plasma membrane is a trilaminar (three layered). Membrane of lipoprotein has shown bimolecular lipid structure sandwiched by two (outer and inner) layers of protein molecules. The electron microscopic studies have confirmed this protein-lipid-protein arrangement in plasma membrane. The plasma membrane of most cells varies from 100^{O}A to 215^{O}A in thickness. The plasma membrane of most cells composed of mainly carbohydrates, lipids and proteins.

A Typical Animal Cell

Molecular structure of-the plasma membrane: The plasma membrane composed of two layers of protein molecules and two layers of lipids molecules. The lipid molecules occurred in chains. In plasma membrane two molecular chains of lipids remain parallel to each other and form a bimolecular or double-layered structure. Both lipid layers remain linked with each other by the inner ends of lipid molecules, which are non-polar and hydrophobic in nature. Both the, layers of lipids are able to be held together due to Vanderwaal's forces at these non-polar ends. The lipid layer is enclosed by outer and inner layers of proteins. The lipid molecules remains linked with the molecules of protein layers by their outer polar and hydrophilic ends. In the hydrogen bonds linkages or electrostatic forces bind the molecules of lipids and proteins together. The carbohydrate molecule occurs in association of protein molecules and provides stability to lipoprotein complex. The protein layer provides elasticity and mechanical resistance to the plasma membrane.



Specialized structures of plasma membrane: The cell surface of certain cells performs various physiological activities such as absorption, secretion, transportation etc. to perform such sort of specialized functions certain modifications are inevitable in the plasma membrane such cells. The most important specialized structures of plasma membrane are microvilli, desmosomes, hemidesmosomos, septate desmosomes, terminal bars, inter-digitations, tight junction gap junction and infoldings.

Primarily the plasma membrane provides mechanical support and external form to the protoplasm and it also delimits the protoplasm from the exterior, checks the entry or exit of undesirable substances and due to semi-permeability it transmits necessary material to and from the cells. Though the plasma membrane is a limiting barrier around the cell but it performs various important physiological functions which are as follows:

Permeability: The plasma membrane is a thin, elastic membrane around the cell which usually allows the movement of small ions and molecules of various substances through it. This nature of plasma membrane is termed as permeability.

Osmosis: The plasma membrane is permeable to water molecules. To and fro movement of water molecules thro' the plasma membrane occurs due to the differences in the concentration of the solutes on its either side. The process by which the water molecules pass through a membrane from a region of higher water concentration to the region of lower water concentration is known as osmosis. The process in which the water molecules enter into the cell is endosmosis, while the reverse process which involves the exit of the water molecules from the cell is known as exosmosis.

Diffusion or passive transport: When molecules of two kinds are placed to mix together they tend with each other by a process known as diffusion. The diffusion of certain solutes or substances takes through the plasma membrane. Such diffusing solute out particles, require no energy for the diffusion process through the plasma membrane. The diffusion of ions through the plasma membrane depends on the concentration and electrical gradients.

Active transport: When the molecules or ions move through plasma membrane from low concentration to higher concentration they require energy for such movement. The energy is provided by adenosine triphosphate (ATP) which produced by oxidative phosphorylation in the mitochondria.

Endocytosis and exocytosis: The plasma membrane participates actively in the ingestion of certain large-sized foreign or food substances. The process by which the foreign substances are taken and digested is known as endocytosis. In the process of exocytosis the cells which have secretory function such as pancreatic cells pass out their enzymatic secretions outside the cell.

According to the nature of the food or foreign substances the endocytosis may be classified into two types, viz., (i) pinocytosis and, (ii) phagocytosis.

(I) Pinocytosis: When the ingestion of fluid material in bulk takes place by the cell through the plasma membrane, the process is-known as pinocytosis.

(II) Phagocytosis: Sometimes the large-sized solid food or foreign particles are taken in by the cell through the plasma membrane. The process of ingestion of large-sized solid substances by the cell is known as phagocytosis.

Cytoplasm: The plasma membrane is followed by the cytoplasm which is distinguished into following structures:

A. Cytoplasmic matrix: The space between plasma membrane and the nucleus is filled by an amorphous, translucent, homogeneous liquid known as cytoplasmic matrix or hypoblasm. The cytoplasmic matrix consists of various inorganic molecules such as water, salts of Na, K and other metals and various compounds, viz., carbohydrates, lipids, proteins, nucleoprotein, nucleic acids (RNA and DNA) and a variety of enzymes. The peripheral layer, of cytopiasmic matrix is relatively non-granular, viscous, clear and rigid and is known is the plasma gel, or cortex or corcoid layer or ectoplasm. The inner portion of cytoplasmic matrix is granular less viscous and is known as endoplasm.

B. Cytoplasmic structures: In the cytoplasmic matrix certain non-living and living structures are suspended. the non-living structures are called paraplasm, deutoplasm or inclusions, while the living structures are membrane bounded and are called organoids or organelles. Both kinds of cytoplasmic structures are as follows

Cytoplasmic inclusions; The cytoplasmic matrix contains many refractive granules of various sizes and shapes known as trophoplasm. These granules in animal cells are known as Cytoplasmic inclusions or deutoplasm or paraplasm. It includes oil drops, yolk granules, pigments, secretory granules and glycogen granules.

Cytoplasmic organelles: the cytoplasmic matrix contains many large sized living structures known as cytoplasmic organelles, which perform various important biosynthesis and metabolic activities such as respiration, storage, synthesis, transportation, support and reproduction. The cytoplasmic organelles are as follows.

Endoplasmic Reticulum (ER): This is a complex network of tubes, the lumen of which is filled with fluid. 2 types of endoplasmic reticula are seen. [1] Tubes with a smooth surface are called smooth ER. They secrete lipids. [2] Tubes with spherical bodies (ribosomes) attached are known as rough ER. The functions of the ER are to form the skeletal framework of the cell, to provide a pathway for the distribution of nuclear material from one cell to the other and to synthesize fats, steroids and cholesterol with the help of enzymes secreted by the cell.

Mitochondria: These may be cylindrical, rod-shaped or spherical and distributed in the cytoplasm. Each mitochondrion is bound by a double membrane. The inner membrane is folded into ridges called cristae, which increase the surface area of the membrane. It is in the mitochondria that the sugar is finally burnt during cellular respiration. The energy thus released is stored as high-energy chemicals called ATP (adenosine triphosphate). Hence, mitochondria are termed as the "power house" or the "power plant" of the cell.

Golgi apparatus: Also known as Golgi Complex or Golgi Bodies, they consist of tiny, elongated, flattened sacs (cisternae), which are stacked parallel to one another along with some vacuoles and clusters of vesicles (*usually found close to the nucleus.*). The function of the Golgi body is to secrete certain hormones and enzymes. It also forms lysosomes and peroxisomes.

Lysosomes: These are tiny, spherical, sac-like structures scattered all over the cytoplasm. Their main function is digestion. They contain powerful destructive enzymes capable of digesting all organic material, and hence called "digestive bags". Lysosomes present in white blood cells are capable of digesting bacteria and viruses. During starvation, lysosomes digest proteins, fats and glycogen in the cytoplasm, and supply energy to the cell. They are also capable of digesting worn out cell organelles, or even digesting the entire damaged cell containing them. Hence, "suicide bag" is a sobriquet that is often used for Lysosomes.

Ribosomes: These are spherical, granular particles which occur freely in the matrix or remain attached to the rough ER. Ribosomes contain RNA (ribonucleic acid) and proteins. Their function is to provide the surface for protein synthesis.

Peroxisomes: These organelles are found in the liver and kidney cells. They are small, membrane-bound sacs, and contain powerful oxidative enzymes.Their chief function is to remove toxic substances.

Centrosome: This is found in the cytoplasm near the outer surface of the nucleus and contains two cylinders called centrioles. The centrosome is found only in the animal cell. The centrosome and the centrioles play an important role by forming the poles of the spindle during cell division.

Plastids: These organelles are found only in plant cells. Plastids are of three types

Chloroplasts: They are green and found in leaves. The green colour is due to the presence of chlorophyll.

Chromoplasts: They are yellow, orange and red, and found in flowers and fruits.

Leucoplasts: They are colourless and found in roots, seeds and underground stems. The function of the chloroplast is to trap solar energy for photosynthesis. Chromoplasts impart colour to flowers to attract insects for pollination. Leucoplasts store food in the form of carbohydrates, fats and proteins.

Nucleus: This is a prominent, spherical or oval structure found at the centre of the cell. It is the controlling centre of all cell activities and has been described as the brain of the cell. It regulates all metabolic and hereditary activities of the cell.

The nucleus is composed as follows: ***[1]*** Nuclear Membrane, ***[2]*** Nucleoplasm, ***[3]*** Nucleolus and ***[4]*** Chromatin network

Nuclear membrane: This is a double-layered membrane which separates the nucleoplasm from the cytoplasm. The nuclear membrane has minute pores which allow the selective transfer of material between the nucleoplasm and the cytoplasm.

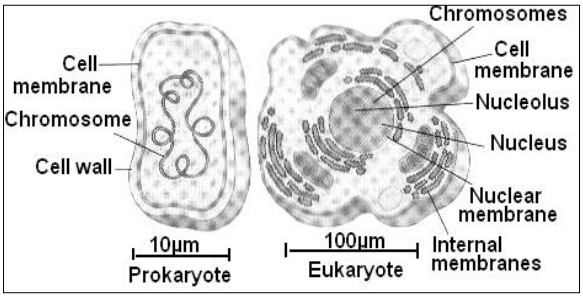
Nucleoplasm: Within the nuclear membrane, completely filling up the space is a clear, semi-solid, granular substance or matrix called the nucleoplasm. The nucleolus and the chromatin network lie suspended in the nucleoplasm.

Nucleolus: This dense, spherical granule found in the nucleus contains RNA (ribonucleic acid) which is responsible for protein synthesis in the cytoplasm.

Chromatin network: These are very fine thread-like, coiled filaments uniformly distributed in the nucleoplasm. At the time of cell division, the chromatin becomes thick and ribbon like and are known as chromosomes. The chromosomes contain genes, which are composed of DNA (deoxy-ribonucleic acid). Genes are responsible for storing and transmitting hereditary characteristics from one generation to another. A gene is the functional unit of a chromosome. Genes are arranged in single linear order along the chromosome. One gene may be responsible for a single characteristic, or a single characteristic may be transmitted by a set of genes.

Prokaryotic and Eukaryotic Cells

The main difference between these two cell types is that Prokaryotic cells do not have a nuclear membrane. The nuclear material consists of a single chromosome and lies in the cytoplasm. The nuclear region in the cytoplasm is called nucleoid. Membrane-bound organelles are absent. Prokaryotic cells are found in bacteria and cynobacteria (blue-green algae).



Differences between Prokaryotic cells and Eukaryotic cells

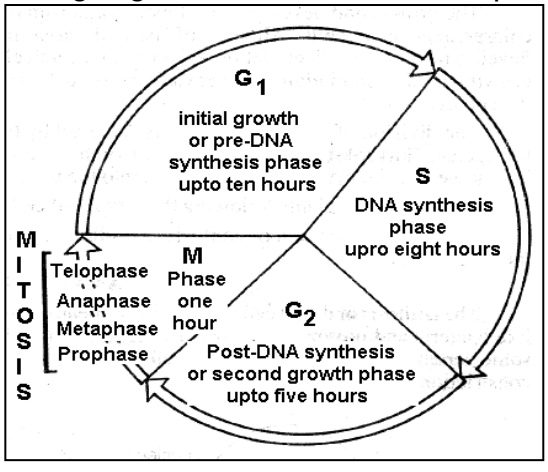
Prokaryotic Cells	Eukaryotic Cells
Very minute in size	Fairly large in size
Nuclear region (nucleoid) not surrounded by a nuclear membrane	Nuclear material surrounded by a nuclear membrane.
Single chromosome present	More than one chromosome present
Nucleolus absent	Nucleolus present
Membrane bound cell organelles are absent	Membrane bound cell organelles are present
Cell division by fission/budding (no mitosis)	Cell division by mitosis or meiosis

MITOSIS:

The mitosis occurs in the somatic cells and it is meant for the multiplication of cell number during embryogenesis and blastogenesis of plants and animals. Fundamentally it is related with the growth of an individual from zygote to adult stage. One of the basic characteristics of mitotic cell division which is meant for growth due to multiplication is that it gives rise to two daughter cells, which resemble each other and also parent cell qualitatively and quantitatively (i. e., the chromosome number of mitotic product cells remain the same- like the parent cell).

The mitosis composed of two apparatuses, viz., chromatic apparatus which includes the chromosomes and the nucleolus and the achromatic apparatus which in its turn includes the centrioles and spindle.

The basic outline of mitosis is the same in all living organisms. It includes four phases namely G1 phase, S phase, G2 phase and mitotic phase which occur in succession and forming the so called cell cycle. The G1 phase, S phase and G2 phase are collectively forming the interphase. Thus, in continuously dividing cells, an individual cell passes through following two main phases of cell or mitotic cycle:



A. Interphase;

B. Mitotic phase.

A: Interphase:

The resting phase or stage between the two mitotic divisions is known as the Intermidiate phase or interphase. In interphase no division occurs but in the nucleus and cytoplasm active metabolic activities occur and also increase in the volume of the cytoplasmic and nuclear substances takes place. The interphase is the longest phase of the mitotic cycle and it takes one or two days in its completion. During the interphase following events take place:

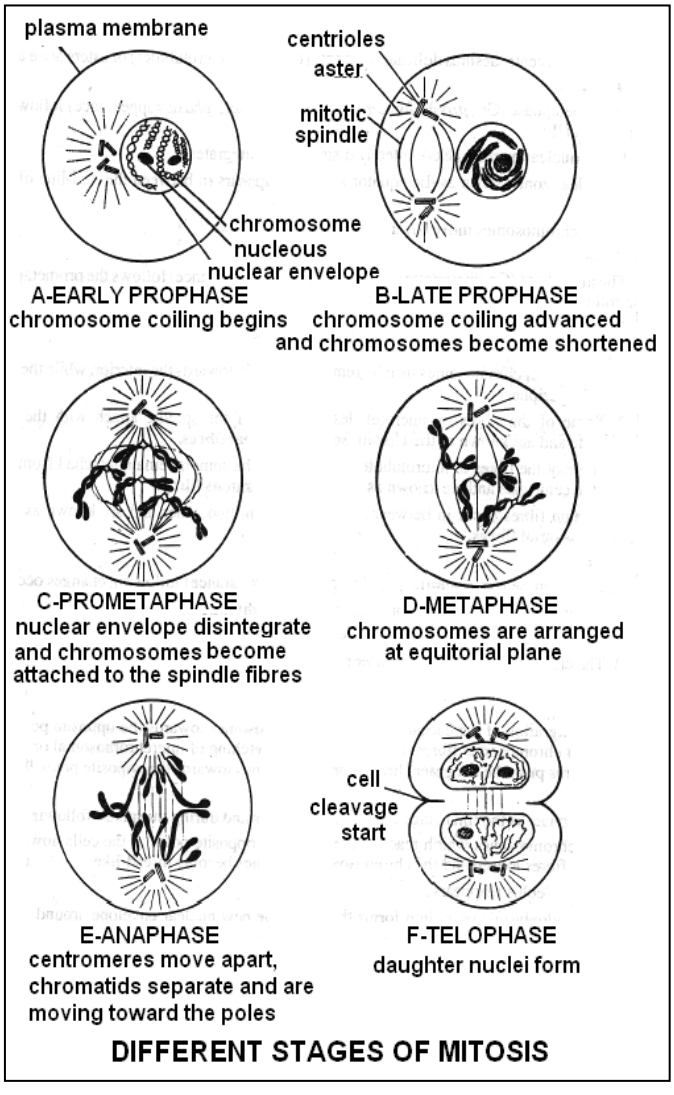
1) The nuclear envelope remains intact.
2) The chromosomes occur in the form of diffused, long, coiled and indistinctly visible chromatin fibres.
3) The DNA amount becomes double.
4) Due to the accumulation of ribosomal RNA (rRNA) and ribosomal proteins the nucleolus attains the maximum size.
5) A daughter centriole is originated near the already existing centriole and, thus, an interphase cell has two centrioles or adiplosome.

B. Mitotic Phase

The main mitotic cell division occurs during mitotic phase or M phase which includes the following phases.

Prophase: The prophase is the actual first phase of the mitosis. During the prophase following events take place in the cell:

1) The cell becomes spheroid, refractive and viscous.
2) The disintegration of nuclear envelope starts.
3) Due to the DNA duplication in the interphase each chromosome now possesses two chromatids. Each clnomatid consists of a single DNA molecule wrapped in the nucleoproteins.
4) Both chromatids remain connected with each other by the centromere and both remain closely associated along their entire lengths.
5) The chromatids become shortened and thickened.
6) The nucleolus starts to disappear.
7) Each centriole separates and migrates towards the opposite poles of the cell. Each centriole duplicates, so that both poles of the cell contain pair:dcentrioles or diplosome.
8) The centrosome forms an elongated body or bridge known as the centrodesmus in between the two centrioles.
9) From the centrodesmus delicate filaments (microtubules) or asters arise and form the spindle.



DIFFERENT STAGES OF MITOSIS

Prometaphase: In prometaphase following changes usually occur in the cell:

1) The nuclear membrane completely disintegrates.
2) A clear zone known as the equator appears in between the mid-line of the spindle and the nucleus.
3) The chromosomes move towards the equator.

Metaphase: the metaphase follows the prometaphase and during this phase following events occur in the cell:

1) Each chromosome reaches to the equator and all arrange themselves radially at the periphery of the spindle.
2) The smaller chromosomes usually remain towards the interior, while the larger chromosomes remain at the periphery.
3) Some of the fibres of microtubules of the spindle attach with the centromere of each chromosome and are known as the chromosomal fibres.
4) Some of the fibres or microtubules of the spindle remain attached from one end to the other end with the centrioles and are known as continuous fibres.
5) Certain fibres occur in between the chromosomes and are known as interzonal fibres or interchromosomal fibres.

Anaphase: In the anaphase following changes occur in the cell :

1) The centromere of each chromosome divides into two.
2) The chromatids of the each chromosome are separated and form two chromosomes.
3) The chromosomes become shorter and thicker and migrate towards the opposite poles of the cell.
4) The migration of the daughter chromosomes towards the opposite poles is achieved by the contraction of chromosomal fibres and the stretching of inter chromosomal or interzonal fibres. The interzonal fibres push the daughter chromosomes towards the opposite poles.

Telophase: The telophase is the final stage of mitosis and during this phase following events occur:

1) The chromosomes which reach at the opposite poles of the cells now elongate, the coils of DNA protein fibres loosen and the chromosomes become thread-like.
2) The nucleolus reappears.
3) The endoplasmic reticulum forms the new nuclear envelope around the chromosomes and the nucleolus.
4) The microtubules of the aster and mitotic spindle rearrange and disappear. Thus, after the telophase two daughter nuclei are formed due to the karyokinesis. The karyokinesis is followed by the cytokinesis.

Cytokinesis: In the process of cytokinesis the cytoplasm splits from the equatorial region and the two daughter halves of the cytoplasm are separated. Soon after a unit membrane of lipoprotein develops in between the two daughter cells. The cytokinesis of animal cells involves the cyclosis of the cytoplasm, formation of a contractile ring, the expansion of the cell membrane, ATP and interaction of the spindle and asters with the cell surface, while in the plant cells the cytokinesis involves the movement of the endoplasmic reticulum and dicytosomes to the equator where they fuse to form the primary cell wall.

Significance of Mitosis:

The importance of the mitosis for the organisms has been summarized as follows points

1) In mitotic division, the chromosome number in each daughter cell remains the same like the parent cell.
2) The mitosis helps the cell in maintaining its proper size.
3) Through the process of the mitosis an equilibrium is maintained in the amount of DNA and RNA contents
4) The mitosis provides the opportunity for the growth and development of organs and the body of the organisms.
5) Due to the mitosis the old decaying and dead cells are replaced by the new cells.
6) The mitosis helps the organisms in the asexual reproduction.
7) The gonads and the sex cells also depend on the mitosis for the increase in their number.

MEIOSIS

The meiotic division is of utmost importance for those organisms in which the union o the haploid gametes takes during the sexual reproduction. By reducing the number of chromosomes of the diploid germ cells into the haploid gametes the meiosis maintains a constant number of the chromosomes in the species. Thus meiosis helps in alternation of generation of haplodic generations of plants and animals. In the process of meiosis the chromosomes divide once and the nucleus and cytoplasm divide twice. Due to the meiosis four haploid cells are formed from the single diploid cell. The process of meiosis is fundamentally same in all the animals and plants but certain biologists recognised following three types of meiotic division according to their occurrence at different stages of the life cycle of the organisms.

1) **Sporogenetic meiosis:** The meiosis occurring at the time of spore formation is referred to as the sporogenetic meiosis. This type of meiosis is commonly found in plants.
2) **Gametic meiosis:** In most animals and lower plants the meiosis occurs at the time of gametogenesis (spermatogenesis and oogenesis) and is known as gametic meiosis.
3) **Zygotic meiosis:** In certain lower plants, sometimes the meiosis occurs immediately after the fertilization of the egg by a sperm. This type of meiosis is known as zygotic meiosis.

Process of Meiosis

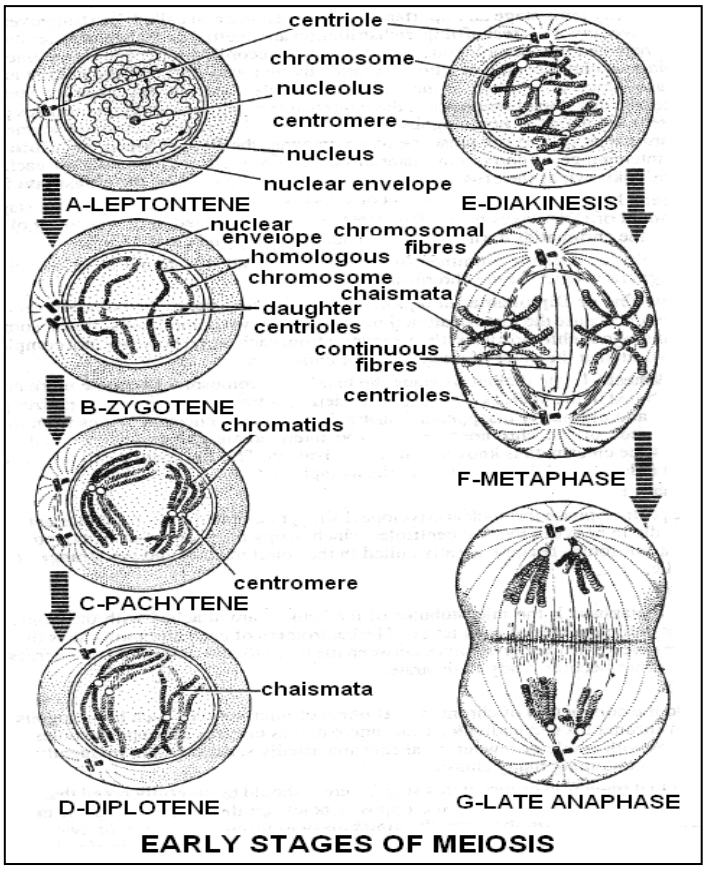
The meiotic division includes two complete divisions of a diploid cell resulting into four haploid nuclei. The first meiotic division includes a long prophase in which the homologous chromosomes become closely associated to each other and interchange of hereditary material takes place between them. Further in the first meiotic division the reduction of chromosome number takes place and, thus, two haploid cells are resulted by this division. The first meiotic division is also known as the heterotypic division. In the second meiotic division, the haploid cell divides mitotically and results into four haploid cells. The second meiotic division is also known as the homotypic division. In the homotypic division pairing of chromosomes, exchange of the genetic material and reduction of the chromosome number do not occur. Both the meiotic divisions occur continuously and each includes the usual stages of the mitosis, viz., prophase, metaphase, anaphase and telophase. The prophase of first meiotic division is very significant phase because the most cytogenetical events such as synapsis, crossing over, etc., occur during this phase. The prophase is the longest meiotic phase, therefore, for the sake of convenience it is divided into six substages, viz., proleptonema (proleptotene), leptonema (leptotene), zygonema (zygotene), pachynema (pachytene), diplonema (diplotene) and diakinesis. The successive meiotic substages can be represented as follows

Heterotypic Division or First mitotic division: In the beginning of the first meiotic division the nucleus of the meiocyte starts to swell up by absorting the water from the cytoplasm and the nuclear volume increases about three folds. This increase in the volume of the nucleus causes modification of nuclear components. After these changes the cell passes to the first stage of first meiotic division which is known as prophase.

First Prophase: The first prophase is the longest stage of the meiotic division. During this stage the amount of DNA becomes double. Most of the synthesis of DNA occurs at the beginning of this phase. It includes following substages:

1) **Preleptotene or Preleptonema:** The preleptotene stage closely resembles with the early mitotic prophase. In this stage the chromosomes are extremely thin, long, uncoiled, longitudinally single and slender thread like structures.

2) **Leptotene or Leptonema:** In the leptotene stage the **chromosomes** become more uncoiled and assume a long thread-like shape. The chromosomes at this stage take up a specific orientation inside the nucleus, the ends of the chromosomes converge toward one side of the nucleus, that side where the centrosome lies (the bouquet stage). The centriole duplicates and each daughter centriole migrates towards the opposite pole of the cell. On reaching at the poles, each centriole duplicates and, thus, each pole of the cell possesses two centrioles or a single diplosome.



EARLY STAGES OF MEIOSIS

3) **Zygotene or Zygonema:** In the zygotene stage, the pairing of homologous chromosomes takes place. The homologous chromosomes which come from the mother (by ova) and father (by sperm) are attracted towards each other and their pairing takes place. The pairing of the homologous chromosomes is known as synapsis. The synapsis begins at one or more points along the length of the homologous chromosomes. Three types of synapsis have been recognised.

   1. **Protemririal synapsis:** In proterminal type of synapsis, the pairing in homologous chromosomes starts from the end and continues towards their centromeres.
   2. **Procentric synapsis:** In procentric synapsis the homologous chromosomes start pairing from their centromeres and the pairing progresses towards the ends of the homologous chromosomes.
   3. **Localized pairing or Random synapsis:** The random type of synapsis occurs at various points of homologous chromosomes. The pairing of the homologous chromosomes is very exact and specific. The bouquet is supposed to maintain a regularity in the synapsis mechanlsm.

4) **Pachytene or Pachynema:** In the pachytene or pachynema-stage, the pair of chromosomes become spirally around each other and cannot be distinguished separately. In the middle of the pachynema stage each homologous chromosome splits lengthwise to form two chromatids. Actually, doubling of the DNA molecule strands, which is necessary for the subsequent duplication of the chromosomes, occurs earlier, before the beginning of meiotic prophase. Through the earlier part of the meiotic prophase however, the DNA molecule in each chromosome behaves as a single body.

In the pachynema stage, this is now changed, the two chromatids contalning of each chromosome half of the DNA present in the chromosome at start, become partially independent of one another although they still continue to be linked together by their common centromere. The pachynema chromosome, thus, consists of four chromatids closely joined together in one complex unit called a bivalent, because it actually contains a pair of chromosomes.

During pachynema stage an important genetic phenomenon called "crossing over" takes place. The crossing over involves reshuffling, redistribution and mutual exchange of hereditary material of two parents between two homologous chromosomes. According to recent views, one chromatid of each homologous chromosomes of a bivalent may divide transversely by the help of an enzymes, the endonuclease which is reported to increase in the nucleus during this stage. After the division of chromatids, the interchange of chromatids segments are united with the chromatids due to the presence of an enzyme, the ligase. This process of interchange of chromatid material between non-sister chromatids of homologous chromosomes is known as the crossing over which is accomplished by Christian formation.

During the pachytene and zygotene stages, synthesis of small amount of DNA takes place, This DNA amount is utilized in the repairing molecules of the chromatids during the chiasmata formation and crossing over. The nucleolus remains-prominent up to this stage and it is formed to be associated with nucleolar organizer region of chromosome.

5) **Diplotene or Diplonema:** In diplotene or diplonema stage, the homologous chromosomes repel each other because the force of attraction between the two homologous chromosomes decreases. The two homologous chromosomes, thus, separate from each other however, not completely because both remain united at the point of interchange or chiasmata.

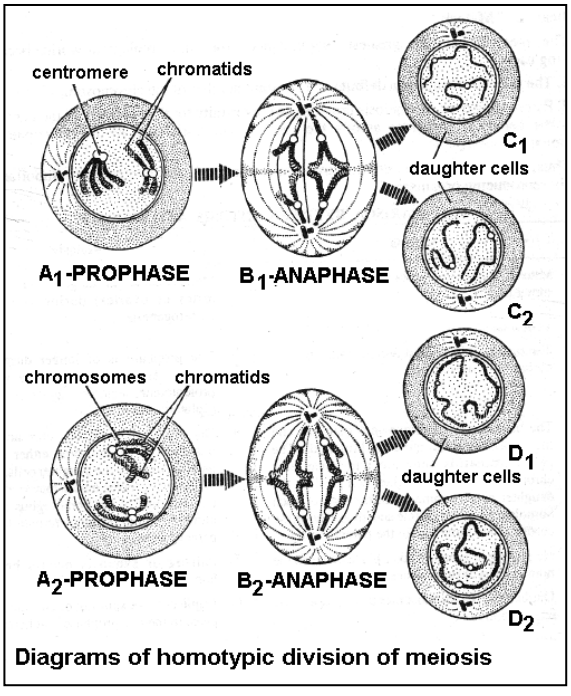
6) **Diakinesis:** In the diakinesis stage the bivalent chromosomes become more condensed and evenly distributed in the nucleus. The nucleolus detaches from the nucleolar portion of the chromosome and ultimately disappears. During diakinesis the chiasma moves from the centromere towards the ends of the chromosomes and the intermediate chiasmatas diminish. This type of movement of the chiasmata is known as terminalization. The chromatids still remain connected by the terminal chiasma and these exist up to the metaphase.

Prometaphase I: in the prometaphase the nuclear envelope disintegrates and the microtubules get arranged in the form of spindle in between the two centrioles which ocupy the position of two opposite poles of the cell. The chromosomes become greatly coiled in the spiral manner and get arranged on the equator of the spindle.

Metaphase I: in the metaphase I, the microtubules of the spindle are attached with the centromeres of the chromosomes of each tetrad. The centromere of each chromosome is directed towards the opposite poles. The repulsive forces between the homologous chromosomes increases greatly and chromosomes become ready to separate.

Anaphase I: Due to the contraction of chromosomal fibres of microtubules each homologous chromosome with its two chromatids and undivided centromere moves towards the opposite poles of the cell. The chromosomes with single or few terminal chiasma usually separate more frequently than the longer chromosomes containing many chiasma. The actual reduction occurs at this stage. Here it should be carefully noted that the homologous chromosomes which move towards the opposite poles are

the chromosomes of either parental or maternal origin. Moreover, because during the chiasma formation out of two chromatids of a chromosome, one has changed its counterpart, therefore, the two chromatids of a chromosome do not resemble with each other in the genetical terms.



Diagrams of homotypic division of meiosis

Telophase I: In telophase I, the endoplasmic reticulum forms the nuclear envelope around the chromosomes and the chromosomes become uncoil. The nucleolus reappears and thus,two daughter chromosomes are formed. After the karyokinesis, cytokinesis occurs and two haploid cells are formed.

Both cells pass through a short resting phase or interphase. In case of Trillium, telophase I & interphase do not occur & anaphase I is followed by prophase II directly.

Homotypic or Second Meiotic Division: The homotypic or second meiotic division is actually the mitotic division which divides each haploid cell into two diploid cells. The second meiotic division includes following four stages:

Prophase II: In the prophase second, each centriole divides into two and, thus, two pairs of centrioles are formed. Each of pair of centrioles migrate to the opposite pole. The microtubules of fibres get arranged in the form of spindle at the right angle of the spindle of first meiosis. The nuclear membrane and the the nucleolus disappear. The chromosomess with two chromatids become short and thick.

Metaphase II: During the metaphase II, the chromosomes get arranged on the equator of the spindle. The centromere divides in to two, thus, each chromosome produces two monads or daughter chromosomes. The microtubules of the spindle are attached with the centromere of the chromosomes.

Anaphase II: Daughter chromosomes move towards opposite poles due to contraction of chromosomesal microtubules and stetching of interzonal microtubule of the spindle.

Telophase II: The chromatids migrate to the opposite poles and now known as chromosomes. The endoplasmic reticulum forms the nuclear envelope around the chromosomes and the nucleolus reappears to synthesis of ribosomal RNA (rRNA) by ribosomal DNA (rDNA) and also due to accumulation of ribosomal proteins.

After the karyokinesis in each haploid meiotic cell the cytokinesis occurs and, thus, four haploid cells are resulted. These cells have different types of chromosome due to crossing over in the prophase I.

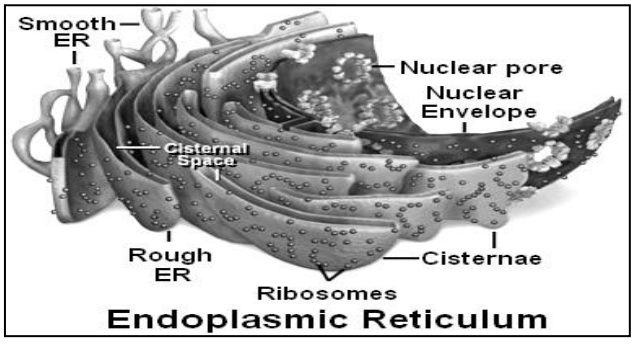
Significance of Meiosis:

1) The meiosis maintains a definite & constant number of chromosomes in organisms.
2) By crossing over, the meiosis provides an opportunity for the exchange of the genes and, thus, causes the genetical variations among the species (evolutionary process).
3) Thus, the meiosis has a peculiar taxonomic, genetical, and evolutionary importance for the sexually reproducing organisms.

COMPARISION BETWEEN MITOSTS AND MEIOSTS

Mitosis	Meiosis
1) Mitosis occurs continuously in the body or somatic cells.	1) Meiosis occurs in the germ cells (the cells of testes or ovaries) during the process of gametogenesis.
2) The whole process completes in one sequence or phase.	2) The whole process completes in two successive divisions which occur one after the other.
Prophase	
3) The prophase is of short duration and does not include any substage.	3) The prophase is of longer duration and it completes in six successive stages, i.e, proleptotene, leptotene, zygotene, pichytene, diplotene and diakinesis.
4) The homologous chromosomes (parental and maternal) duplicate into two chromatids. The two chromatids separate and form new chromosomes. Each daughter cell receives the daughter chromosome or chromatids of each homologous chromosome and, thus, having the chromosome number likes the parental cells.	4) Out of two homologous chromosomes only one type of chromosome either maternal or parental move to the daughter cells. A daughter cell, thus, receives only a maternal or paternal chromosome of the homologous pair and the number of chromosomes remain half than the parental cells.
5) No pairing or synapsis takes place between the homologous chromosomes.	5) Pairing or synapsis occurs between the homologous chromosomes.
6) Duplication of chromosomes takes place in the early prophase.	6) Duplication or splitting of chromosomes takes place in the late prophase (pachytene stage).
7) No chiasma formation or crossing over takes place.	7) Chiasma formation or crossing over takes place.
8) The exchange of the genetic material between the homologous chromosomes does not occur.	8) The exchange of the genetic material takes place between the non-sister chromatids of homologous chromosomes.
Metaphase	
9) The chromatids occur in the form of dyads.	9) The chromatids of two homologous chromosomes occur as the tetrads.
10) The centromeres of the chromosomes remain directed towards the equator and the arms of the chromosomes remain directed towards the poles.	10) The centromeres of the ctnomosomes remain directed towards the poles and the chromosomal arms remain directed towards the equator.
Anaphase	
11) The chromosomes are the monads, i.e., having single chromatid.	11) The chromosomes are the dyads, i.e., having two chromatids and single centromere.
12) The chromosomes are long and thin.	12) The chromosomes are short and thick.
13) The telophase always occurs.	13) The first telophase is sometimes omitted.
Significance	
14) In mitotic division the chromosome number in each daughter cell remains the same like the parent cell.	14) In meiotic division the chromosome nurnber is reduced to half in the daughter cells than the parental cells.
15) A diploid cell produces two diploid cells by a mitotic division.	15) A diploid cell produces four haploid cells by a meiotic division.

Endoplasmic Reticulum (*ER*): The occurrence of the ER varies from cell to cell. The egg and embryonic cells lack in endoplasmic reticulum, the spermatocytes have poorly developed ER. The adipose tissues, brown fat cells and adreno-cortical cells of opossum's testis contain only smooth type of ER.



Endoplasmic Reticulum

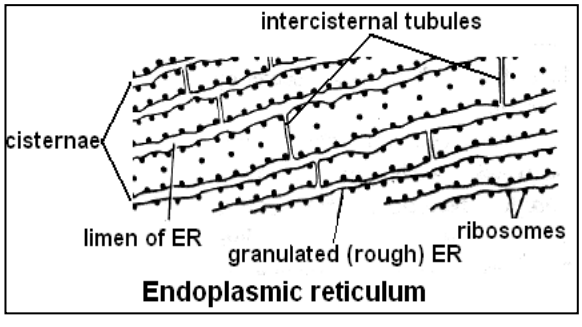
The cells of those organs which are actively engaged in the synthesis of proteins, as the pancreas and other endocrine glands, are found to contain highly developed ER and most ER is of granular type. The liver cells possess both types of ER.

Structure:- The cytoplasmic matrix is traversed by a complex network of inter-connecting membrane bound vacuoles or cavities. These vacuoles or cavities often remain concentrated in the endoplasmic portion of the cytoplasm, therefore, known an ER. This is a complex network of tubes, the lumen of which is filled with fluid.

The endoplasmic reticulum (***ER)*** is composed of the following three kinds of structures, namely, Cisternae, Vesicles, and Tubules. The cavities of cisternae, vesicles and tubules of the ***ER*** are bounded by a thin membrane of 50 to 60Ao thickness. The membrane of endoplasmic reticulum is trilaminar like the unit membrane of the plasma membrane, nucleus, Golgi complex, etc., the sandwiching two thin and transparent layers of phospholipids. The cavity of the endoplasmic reticulum is well developed and acts as a passage for the secretory products.

Cisternae: The cisternae are long, flattened, sac-like, unbranched tubules having the diameter of 40 to 50 micro meter. They remain arranged parallel in bundles or stakes.

Vesicles: The vesicles are oval, membrane-bound vacuolar structures having the diameter of 25 to 500 micro meter. They often remain isolated in the cytoplasm and occur in most cells but especially abundant in the pancreatic cells.



Endoplasmic reticulum

Tubules: The tubules are branched structures forming the reticular systems along with the cisternae and vesicles. They usually have the diameter from 50 to 190 micro meters and occur almost in all the cells.

Types of Endoplasmic Reticulum: Two types of endoplasmic reticulum have been observed in same or different types of cells which are as follows:

Agranular or smooth endoplasmic reticulum (SER):- This type of endoplasmic reticulum possesses smooth walls because the ribosomes are not attached with its membranes. The smooth type of endoplasmic reticulum occurs mostly in those cells, which have no active participation in the synthesis of proteins. The smooth endoplasmic reticulum is generally found in adipose cells, interstitial cells, and glycogen storing cells of the liver, spermatocytes and leucocytes. The muscle cells are also rich in smooth types of ER and here it is known as **sarcoplasmic reticulum.**

Granular or rough endoplasmic reticulum (RER):- The granular or rough type of endoplasmic reticulum possesses rough walls because the ribosomes remain attached with its membranes. Ribosomes play a vital role in the process of protein synthesis. The granular or rough type of endoplasmic reticulum is found abundantly in those cells which are active in protein synthesis such as pancreatic cells, plasma cells, goblet cells, and liver cells. The granular type of endoplasmic reticulum takes basophilic stain due to its RNA contents or ribosomes. The region of the matrix containing granular type of endoplasmic reticulum takes basophilic stain and is named as **ergastoplasm**, **basophilic** bodies, **chromophilic** substances or **Nussle** bodies by early cytologists.

Functions of Endoplasmic Reticulum: The functions of the endoplasmic reticulum are to form the skeletal framework of the cell, to provide a pathway for the distribution of nuclear material from one cell to the other and to synthesize fats, steroids and cholesterol with the help of enzymes secreted by the cell. The endoplasmic reticulum acts as secretory, storage, circulatory and nervous system for the cell. It performs the following important functions:-

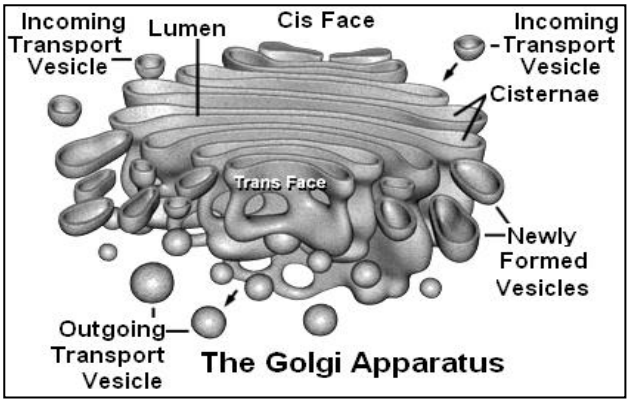
Common functions of granular and agranular endoplasmic reticulum: - The endoplasmic reticulum provides an ultra structural skeletal framework to the cell and gives mechanical support. The exchange of molecules by the process of osmosis, diffusion and active transport occurs through the membranes of endoplasmic reticulum. The endoplasmic reticulum provides increased surface for various enzymatic reactions.

Functions of granular endoplasmic reticulum: - The granular endoplasmic reticulum consisting ribosomes on it will synthesize new proteins (haemoglobin, fibrous proteins, serum proteins, enzymatic proteins etc.), according to the direction of the nuclear DNA.

Functions of agranular endoplasmic reticulum: - The agranular or smooth type of endoplasmic reticulum synthesizes and stores various substances such lipids, glycogen and other synthetic functions.

Golgi body: Golgi body varies in size and form in different cell types but usually has similar organization for any kind of cells. For e.g., well developed in secretory and nerve cells, what is rather small in muscle cells? The Golgi body appears as a coarse network under a light microscope.

Electron microscope shows it as a central stack of flattened sacs or cisternae and many peripheral tubules and vesicles in Golgi body structure.



Cisternae: the cisternae vary in number from 3 to 7 in most anomaly cells and 10 to 20 in plant cells. They are usually equally spaced the pile so that they are nearly parallel to one another, having 200- 300 A˚ wide intracisternal spaces in between. In certain cells the intracisternal spaces contain a layer of parallel fibers called intracisternal elements. The cisternae may be flat but are often curved, having a distinct polarity with a convex face towards the cell membrane and concave face toward the nucleus. They are free of ribosomes and have swollen ends. They look like the smooth ER and are continues with it at certain places. This suggests that the Golgi body is deriving from the smooth ER.

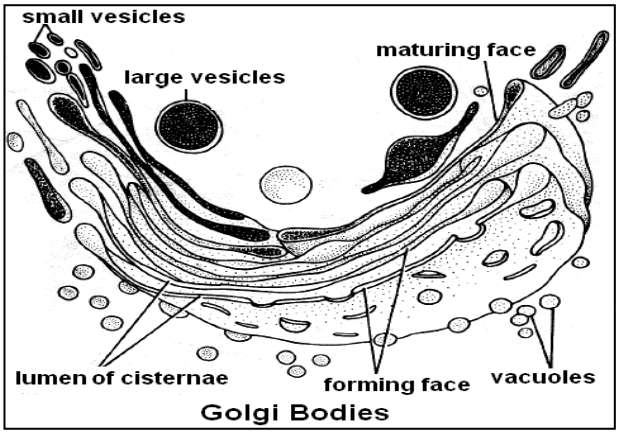
Tubules: They are sort; anatomizing tubules arise from the periphery of the cisternae. Some of these enlarge at their ends to form vesicles.

Vesicles: lie near the ends and concave surface of the Golgi body. They are pinched off from the tubules of the cisternae. They are of 2 types: smooth and coated.

Smooth vesicles: They have smooth surface and contain secretions of the cells. They are also called secretory vesicles. They arise from the ends of cisternal tubules.

Coated vesicles: It have rough surface they also arise from the cisternal tubules.

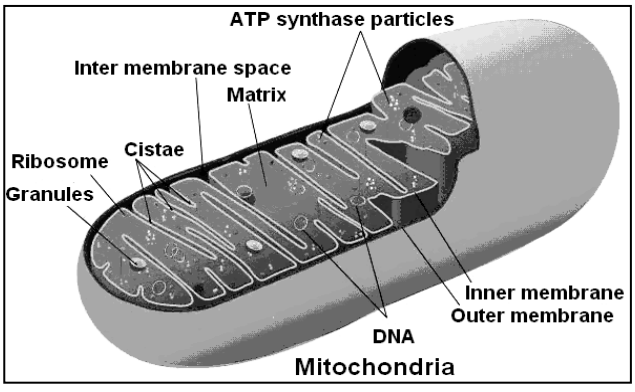
The Golgi body structure has three function regions: cis region that lies nearest the ER, medial region in the middle, and Tran's region with trend Golgi reticulum nearest the plasma membrane. These regions have different enzymes which introduce different modification to secretary and membrane protein passing through them.



Function of Golgi complex: Golgi complex is actively involved in the metabolism of cells. It performs various important functions in each cell.

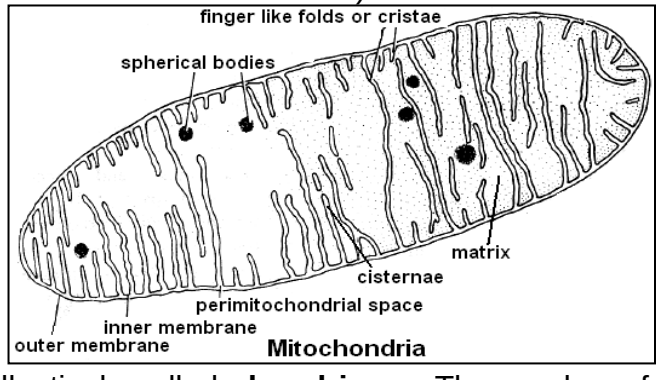
1. This is the primary function of golgi complex. It is basically coenered with the formation of secretory vesicles or primary lysosomes.
2. It acts as packing and forwarding center of cell, reason it functions as condensation unit of cell. At this region molecule of lipid, enzymes, hormones etc. get condensed.
3. Synthesis of carbohydrates: complex carbohydrates are synthesized from simple sugars in golgi complex. Afterword these carbohydrates get combined with proteins to form glycoproteins.
4. Formation of Acrosome: it manufactures acrosomes in testes of vertebrates. Acrosome forms a cap like structure at the head of the spermatozoa, lysosomes contain various enzymes, required for the penetration of vitelline membrane of egg.
5. Production of hormone: in the cells of endocrine glands Golgi complex is avtively concerned with synthesis of various hormones.
6. Formation of Lysossomes: the cisternae of the golgi complex in turn form vesicles (the primary lysosomes) by budding. The primary lysosomes fuse with pinocytic vesicles or with the autophase vesicles and form secondary lysosomes.
7. Formation of cell wall: the cell walls of plant cells are made up of fibrils which contain Polysaccharide along with some lipids and proteins. During cytokinesis a cell plate is formed between the two daughter nuclei and has around or a membrane which later becomes the plasma membrane of daughter cells. There is clear evidence that the polysaccharides are formed in Golgi complex and treansferred to the new cell wall.
8. Formation of Plasma membrane: Golgi complex plays active role in the formation of conjugated compounds, such as phospholipids and lipoproteins,are most essential for the synthesis of plasma membrane. Golgi complex also plays an important part in the synthesis of carbohydrate compounds of the plasma membrane.

Mitochondria: Mitochondria is the main organelles which are the sites of cellular respiration, a catabolic oxygen-requiring process that uses energy extracted from organic macromolecules to produce ATP. Mitochondria are found in nearly all eukaryotic cells. Number of mitochondria per cell varies and is depends on cell's metabolic activity. Mitochondria Is about 1 mm in diameter and 1-10 mm in length. Mitochondria have dynamic structures that move, change their shape and divide.



Structure of Mitochondria: Mitochondria are hollow, sac like cell organelles present in all eukaryotic cells (except mature RBCs of mammals). These are absent from all prokaryotes.

Mitochondria are approximately of the size of bacteria, being 0.2 µm to 1.00 µm in width and 1.00 µm to 10 µm in length. Their diameter does not exceed 1 µm. Thus, they are visible under a light microscope, through their detailed structure could be revealed by electron microscope.

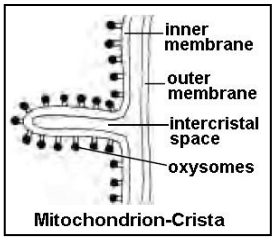


The mitochondria present in a cell are collectively called **chondriome.** The number of mitochondria per cell varies considerably (50- several thousands) depending upon the type of organism and of the cell.

Mitochondria are variable in shape (thread – like/ ovoid/ ellipsoidal/ spherical). Each mitochondrion consists of a fluid-filled cavity, surrounded by two membranes (envelop), enclosing a central fluid mass – the matrix. Each membrane is trilaminar lipo-proteinaceous unit membrane.

The outer membrane is separated from the inner membrane by a space 6-10 nm wide (the perichondrial space or outer chamber). It is more permeable to small molecules and contains some enzymes. However, it is poorer in proteins as compared to inner membrane.

The inner membrane is folded or projected into a number of fingers - like structures called **cristae.** It is more selective in permeability but is richer in enzymes and carriers. Cristae may be branched or unbranched. Some sessile particles attached to the outer membrane are known as **subunits of Parson.**



A number of knob like stalked structures arising from the inner surface of the inner membrane project into the matrix, are called **elementary particles** or **F_1 particles.**

Each oxysome consists of **a head piece, stalk and a base.** The head piece is associated with ATP synthesis and processes the coupling enzyme ATP-ase. The ATPase participates in the final step of **oxidative phosphorylation.** At the base of the particle, extending through inner membrane, are the components of reparatory chains.

The wide space enclosed by inner membrane is inner chamber filled with a dense fluid (the mitochondrial matrix). The matrix contains a highly concentrated mixture of hundreds of different enzymes. All the enzymes of the Krebs cycle and electron transport system – flavoproteins, succinic dehydrogenase, cytochrome b, c, c_1, a and a_3 are embedded in the inner mitochondrial membrane

Function of Mitochondria: Mitochondria are called '**power house'** or '**power plant'** or '**energy converting organelles**' of eukaryotes. It is in these organelles that respiration (transformation of chemical energy into utilizable form) takes place. The food material is gradually oxidized and energy generated is stored in the form of ATP.

The mitochondrion is a power plant and industrial park of the cell where energy stored in the bonds of carbohydrates is converted to a form more useful to the cell (ATP) and certain essential biochemical conversions of amino acids and fatty acids occur. A cell uses energy to synthesize cell-specific materials that it can use for activities such as growth, reproduction, and movement. Energy is transformed from one form to another in mitochondria found in all eukaryotic cells.

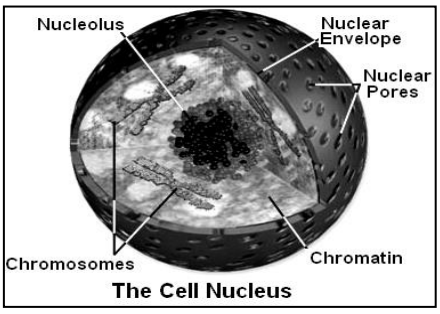
Mitochondrial matrix also contains **mitochondrial DNA** (a circular double stranded DNA molecule). Each mitochondrion has 2-6 DNA circles which carry enough information for the synthesis of about 30 proteins. But this is not enough to build all the proteins required to make a new mitochondrion.

Therefore, mitochondrion has to depend upon nuclear DNA, cytoplasmic enzymes and other molecules supplied by cells.

Functions of mitochondria are listed below:

1. The major function of the mitochondria is to produce energy. The energy giving food molecules are sent to the mitochondrion where they are further processed to produce charged molecules that combine with oxygen and produce ATP molecules. This total process is known as oxidative phosphorylation.
2. NADH and FADH2 from glycolysis, pyruvate oxidation, and the citric acid cycle are oxidized by the respiratory chain, regenerating NAD+ and FAD. Most of the enzymes and other electron carriers of the chain are part of the inner mitochondrial membrane. Oxygen is the final acceptor of electrons and protons, forming water.
3. A Chemiosmotic Mechanism Produces ATP As electrons pass through the series of protein complexes in the respiratory chain, protons are pumped from the mitochondrial matrix into the intermembrane space. As the protons return to the matrix through ATP synthase, ATP is formed.
4. Mitochondria help to maintain proper concentration of calcium ions within the various compartments of the cell.
5. Mitochondria stores calcium.
6. Mitochondria help in the formation of blood components and hormones such as testosterone and estrogen.
7. Mitochondria in the liver help to detoxify ammonia.
8. Mitochondria help in the regulation of membrane potential, cell proliferation and cell metabolism. Production of heat.
9. Mitochondria cause apoptosis or programmed cell death.
10. Mitochondria help in the biosynthesis of heme and steroids.

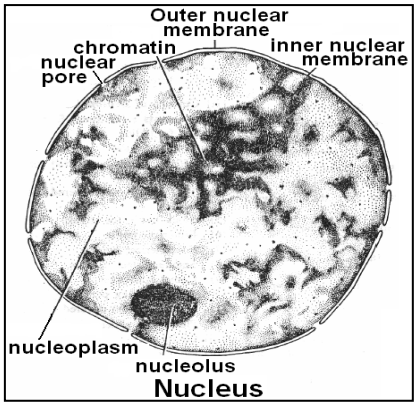
Nucleus: The most prominent structure in the living cells whose presence and absence brings out key difference between two forms called as prokaryotes and eukaryotes. Prokaryotes lack true nucleus structure and considered under developed when compared to eukaryotes with true nucleus structure and are rated as developed. Nucleus is known to be the prominent cell organelle in eukaryotes where in it helps in protection of genetic material called DNA and represents as the site for key activities like replication and transcription. All human cells possess nucleus with the exception of mature erythrocyte which lacks nucleus. Erythrocytes are the key components of bone marrow and possess pigment called hemoglobin.



The Cell Nucleus

Structure: Nucleus is the most prominent, spherical shaped organelle present in every eukaryotic cell. Which is separated from cytoplasm by presence of double membrane layer called nuclear membrane.

The nuclear membrane is with perforations called nuclear pores which help in transportation of proteins and other molecule to cytoplasm. Microscopic observation revealed that nucleus has diameter of 1-3µm and is known that nucleus is mostly in the center. It stains deeply with dyes which are basophilic in nature; contain darkly stained regions called heterochromatin along with lightly stained regions as euchromatin.



Nucleus

It is found that the appearance of nucleus differs as the cell starts dividing. Once cell enters mitotic phase the nucleus structure undergoes disorganization and gets packed and the genome is enclosed in rod like structures called chromosomes.

Structure of Nucleus Function: Nucleus consists of following parts:

1) Nuclear membrane
2) Encloses the contents of Nucleus
3) Double layered
4) Perinuclear space present between two layers.
5) Nuclear pores present on membrane connects them to rest of cell
6) Outer nuclear membrane connected to cytoplasm.

Chromosomes

1) Composed of Chromatin (Long thread like structures that consist of DNA and histones (Protein entities))
2) Chromatin may be of two types based on function:
3) Heterochromatin- Highly condensed, trancriptionally inactive form
4) Euchromatin-less condensed, trancriptionally active

Nucleolus

1) Dense, spherical shaped structure present inside the nucleus
2) Indirect role in protein synthesis by producing ribosomes
3) Disappears during cell division

Components of Cell Structure Nucleus

Nuclear envelope: As cited above the nuclear envelope is with perforations or pores and outer membrane is continued as membranes of rough endoplasmic reticulum. The inner nuclear membrane contains filaments connected to chromatin.

Chromatin: The nucleoplasm in the nucleus contains fibrous material called chromatin fibers which are around 20 nm diameter and have nucleosomes.

Nuclear matrix: The cell structure of nucleus is well controlled by the matrix which is present in between nuclear envelope and chromatin.

Nuclear membrane: The nuclear membrane is defined as the double layered membrane of the nucleus of a cell that separates the nucleoplasm from the cytoplasm that has been permeable to certain molecules.

It is also defined as the double membrane of the nucleus that encloses the DNA and any other genetic material in the eukaryotic cell is called as the nuclear membrane. The basic difference or identifying feature of prokaryotes and eukaryotes is that eukaryotes have a nuclear membrane while prokaryotes are deprived of it.

Components of a Nuclear membrane: The nuclear membrane contains three structural components. These include:

1) Outer nuclear membrane,
2) Inner nuclear membrane,
3) Nuclear pore.

The nuclear envelope is perforated by hundreds of nuclear pore complexes. Around 1,000 - 10,000 nuclear pore are found in nuclear membrane. Nuclear membrane on its periphery has nuclear lamina which has "lamines protein" that provides stability to nucleus. If it collapses, nuclear membrane also collapses so does the nucleus. This happens when karyokinesis or cell division takes place.

Function of Nucleus: The central controlling unit of cell is nucleus. This controlling unit carries the hereditary information to be carried from one generation to another. And the membrane enclosing this important organelle which carries information for next generation is the nuclear membrane. Nucleus is known to have DNA in form of linear arrangement of genes. Genes represent the characteristic feature of cell. Chromosomes indicate collection of genes and nuclear proteins. Nucleus is also known to contain human karyotype indicating 46 chromosomes.

1) Regulates all the Cell activities.
2) Storage of hereditary material (Genes—DNA—Chromatin)
3) Storage of proteins and RNA in nucleolus.
4) Site of Replication and Transcription (Central Dogma of Life)
5) Information stored for synthesis of all proteins found in individual
6) Production of Ribosomes in Nucleolus.
7) Selective transportation of regulatory factors & energy molecules thro' nuclear pores

Function of the Nuclear membrane: The nuclear membrane surrounds the nucleus that is covered with pores and it controls nuclear traffic and the nuclear membrane holds the nucleus together.

1) The nuclear membrane encloses the nucleus of the cell that controls the things which enters and leaves the nucleus. So it is also called as the nuclear envelope. Any material entering into the nucleus through nuclear pore should have a signal called "nuclear localizer signal" (NLS). This signal which brings the material inside the nucleus is called "importin signal" and protein which transports is called "nuclear importin protein". Similar is the case for "exportin signal" and "nuclear exportin protein" which transports material out of the nucleus.

2) The nuclear membrane pores regulate the exchange of materials between the nucleus and the cytoplasm. Inside the inner membrane the nuclear lamina forms a network of filaments which play an important role in mitosis and meiosis.

3) In some eukaryotes a closed mitosis takes place in which the chromosome remains within the nuclear membrane. Here the membrane itself undergoes a division as like the two daughter cells divide.

4) It separates the nucleoplasm from the cytoplasm and the nuclear membrane ensures that the inside of the nucleus is isolated from the cell's cytoplasm which allows two different environments to be maintained.

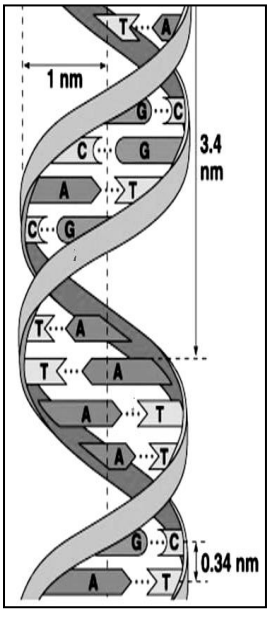
During prophase in the mitotic cell division, the nuclear membrane disintegrates and forms the chromosomes. At the end of Meta phase the nuclear membrane is not present and releases the nuclear lamina.

This is how nuclear membrane plays an important role in maintaining the stability of central controlling unit of cell called "nucleus".

Types of DNA and RNA:

Living Organisms are very complex. All the activities of the body are carried out by hundreds of proteins inside every cell. These proteins specified for a particular function is controlled by a set of molecules called nucleic acids. Nucleic acids are very large molecules made up of a sugar backbone, phosphate molecule and nucleotide base. The genetic information of every living organism is stored inside these nucleic acid molecules. There are two types of nucleic acids namely:

- ✓ **DNA- *Deoxyribonucleic***
- ✓ **RNA - *Ribonucleic acid.***



DNA *(Deoxyribonucleic acid):* In most living organisms (except for viruses), genetic information is stored in the form of DNA. DNA is present in the nucleus of every cell. The nucleotide bases present in the DNA are adenine (A), cytosine (C), guanine (G), and thymine (T).The original structure of the DNA molecule is a double helix. DNA is a double stranded structure. Deoxyribonucleic Acid (DNA) is a long polymer of deoxyribonucleotides. The length of DNA is defined as the number of nucleotides present in it.

Structure of DNA:

1) DNA is formed of number of nucleotides units.
2) Each nucleotide has a nitrogenous base, a pentose sugar and inorganic phosphate.
3) Nitrogen basis occur in DNA belonging to two groups, purine and pyrimidine.

4) DNA has two double ring of purines (Adenine-A and Guanine-G) and two single ring pyrimidine (Cytosine-C and Thymine-T)
5) A nitrogenous base is linked to the pentose sugar through N-glycosidic linkage to form a nucleoside.
6) When a phosphate group is linked to 5 –OH of a nucleoside through phosphodiester linkage, a corresponding nucleotide is formed.
7) The two chains are held together by two hydrogen bonds between adenine-A with Thymine-T and by three hydrogen bonds between Guanine-G with Cytosine-C.
8) The two DNA chains are antiparallel that is, they run parallel but in opposite directions. In one chain the direction 5'→3' while in the opposite one it is 3'→5'.

Double Helical Model and Structure of DNA: In 1953 James Watson and Francis Crick proposed the Double Helix model for the structure of DNA based on the X-ray diffraction data produced by Maurice Wilkins and Rosalind Franklin.

DNA is double helix and formed of two polynucleotide chains which are coiled with one another in a spiral. The nucleotides in a polynucleotide chain are linked together by phosphodiester bond. The two chains of DNA have anti parallel polarity 5' -> 3' in one and 3' -> 5' in other.

Nitrogen bases of two polynucleotide chains form complementary pairs, A opposite to T & G opposite to C. The helix has a constant diameter of 20Ao (2nm) throughout its entire length. The pitch of helix in 3.4nm (34Ao) with roughly 10base pairs in each turn. The average distance between base pairs comes to about 0.34nm.

Four features summarize the molecular architecture of the DNA molecule:

⇨ It is a double-stranded helix.
⇨ It has a uniform diameter.
⇨ It is right-handed (that is, it twists to the right, as do the threads on most screws).
⇨ It is antiparallel (the two strands run in opposite directions).

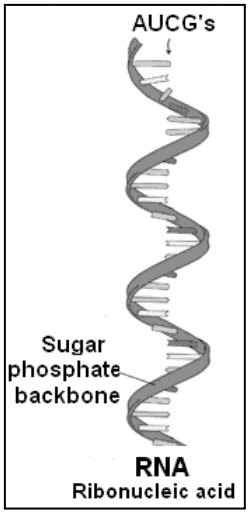
Types of DNA: There are two major types of DNA- Genomic DNA and Mitochondrial DNA;

Genomic DNA / Nuclear DNA: This comprises the genome of an organism. This genomic DNA is spread across 46 chromosomes leading to an expression of genetic traits. The genomic DNA controls expression of the various traits in an organism. The genomic DNA was sequenced as part of the Human Genome Project to study the various functions of the different regions of the genome. Usually, during DNA replication there is a recombination of genes bringing about a change in sequence leading to individual specific characteristics. This way the difference in sequence could be studied from individual to individual. This is very useful in the study of congenital abnormalities. However, any deviation from the normal DNA regulation will lead to Malignancies and other disorders.

Mitochondrial DNA: The DNA located in Mitochondria is called Mitochondrial DNA. mtDNA being derived from the circular bacterial genomes-hence mtDNA is a double stranded circular molecule. mtDNA is always Maternally inherited. Mutations in the mtDNA can lead to maternally inherited diseases. Each Mitochondrion contains about 2-10 mtDNA molecules. Unlike Nuclear DNA which during the process of inheritance undergoes recombination, mtDNA does not change from parent to offspring.

Functions of DNA

- ✓ DNA contains the genetic instructions which are essential for the development and functioning of all living organisms including some viruses.
- ✓ The main function of DNA is the long-term storage of genetic information.
- ✓ DNA is acts and functions as the blue print because it contains the instructions needed to construct other components of cells like RNA and protein molecules.
- ✓ The DNA segments that carry this genetic information are called genes, but other DNA sequences have structural purposes, or are involved in regulating the use of this genetic information. The information carried by DNA is held in the of pieces of DNA called genes.
- ✓ Transmission of genetic information in genes is achieved via complementary base pairing. For example, in transcription, when a cell uses the information in a gene, the DNA sequence is copied into a complementary RNA sequence through the attraction between the DNA and the correct RNA nucleotides. Usually, this RNA copy is then used to make a matching protein.

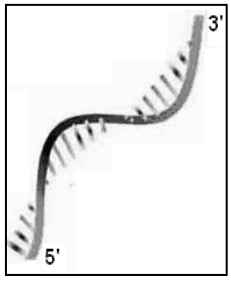


RNA *(Ribose Nucleic acid)***: RNA** is a biologically important molecule abbreviated as "**Ribose Nucleic Acid**". Ribonucleic acid forms the genetic material in smaller organisms namely viruses. RNA is important in the production of proteins in living organisms.

RNA can move around in the cells of living organisms and serves as a genetic messenger, passing the information stored in the cell's DNA from the nucleus to other parts of the cell for protein synthesis.

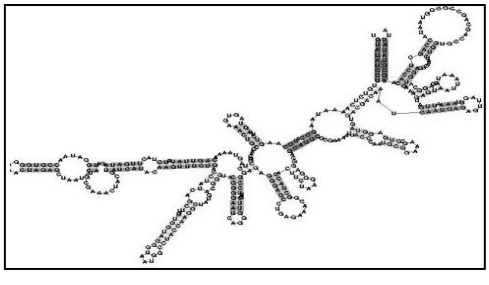
The 4 bases found in RNA are adenine (A), cytosine (C) and guanine (G), RNA does not contain thymine, and instead, the fourth nucleotide present in RNA is the base uracil (U) .RNA is a single stranded structure.

Types of RNA: There are 3 different types of RNA;



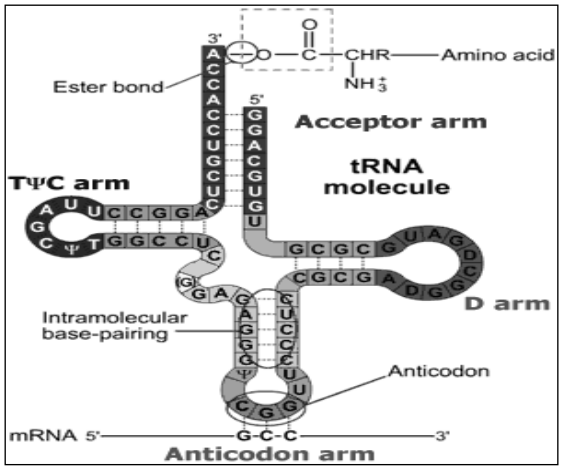
mRNA- Messenger RNA: This carries information from the nucleus to the ribosomes which are sites for protein synthesis. The coding sequence on the mRNA determines the amino acid sequence in the protein. The mRNA is a straight molecule extends from the 5' to 3' end. It is transcribed from a DNA template. On the mRNA nucleotides are arranged into codons consisting of 3 bases each. Each such codon specifies an amino acid.

rRNA – Ribosomal RNA: This is the Ribosomal RNA. The rRNA is synthesized in the nucleolus. In the cytoplasm, ribosomal RNA and protein combine together to form a nucleoprotein called a ribosomes. The ribosomes and mRNA bind to carry out protein synthesis.rRNA is very abundant in the cell and forms about 80% of the total RNA.



The ribosomal RNAs form two subunits namely; the large subunit and small subunit. The Eukaryotic cells have 4 different types of rRNA namely; 28S rRNA, 18S rRNA, 5.8S rRNA and 5S rRNA.

These different rRNA occur in the different ribosomal subunits depending on whether Eukaryotic or Prokaryotic.



tRNA – Transfer RNA: This RNA type is a small chain of about 80 nucleotides. As the name suggests, tRNA transfers specific amino acid molecules to a growing polypeptide chain. The tRNA has a clover leaf model with 5 arms each with a specific function. The tRNA also has anticodon regions that can base pair with the codon region on the mRNA.

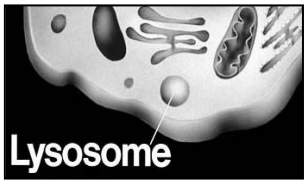
Functions of RNA:-

mRNA: - (Messenger RNA): Messenger RNA is also known as 'Chemical blue print' for the production of proteins. mRNA is transcribed from the DNA template .and carries the coding informations to the site of protein synthesis. t is the RNA which helps to carry the informations produced by the DNA during protein synthesis in all higher organisms such as humans, animals etc

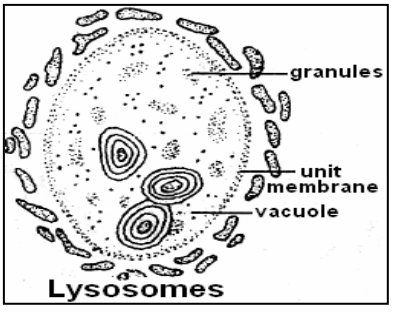
rRNA: - (Ribosomal RNA): Ribosomal RNA is the central component of ribosomes. The important function of rRNA is to decode the mRNA to form amino acids and then relate to tRNA during translation. It catalysts the ribosomal activity during translation phase of protein synthesis such as humans, animals etc.

tRNA: - (Transfer RNA): Transfer RNA is a small RNA molecule that transfers the specific amino acid to the polypeptide chain for the elongation process in the Ribosomes. Each tRNA can be attached to only one amino acid. It helps in the transferring the amino acid from its pool to the site where the production of polypeptide chains take place inside the ribosome such as humans, animals etc

Lysosomes: Lysosomes are single membrane bound, vesicular cell organelles containing hydrolytic enzymes. Lysosomes are formed from ER or Golgi bodies. It is scattered in the cytoplasm. They are found more in WBC, histiocytes of connective tissue, reticuloendothelial cells of liver and spleen. Their size varies from 0.2- 0.8µm.



Structure of Lysosomes: The cytoplasm of both plant and animal cells contain many tiny regular shaped membrane bound vesicles known as lysosomes. Structurally Lysosomes look like membrane bound vesicles or vacuoles (*In animal cells usually spherical*) and measure 0.25 µm in diameter without any characteristic shape or size. Under electron microscope structure of lysosomes appears dense & finely granular & bounded by unit membrane. The unit membrane is trilaminar, consist of protein-lipid-protein. Lysosome contains globules of protein and lipid and carries at least 50 types of hydrolytic enzymes. Lysosomes of injured or dead cells rupture together and release enzymes which lyse the useless cells. They are polymorphic in nature particularly regarding the particle size and irregularities of its internal structure.



This polymorphism suggests that lysosomes are highly dynamic in nature and lights up on what is the lysosomes function. Polymorphism is caused by association of different materials with the primary lysosome.

Classification of Lysosomes: Lysosomes are classified into three types:-

1. **Primary lysosomes:** The newly produced lysosomes are called primary lysosomes, which is a virgin particle in that its digestive enzymes not yet taken part in hydrolysis.
2. **Secondary lysosomes:** The lysosome that is formed by fusion of primary lysosome and phagosomes is termed secondary lysosomes. They are of two types-
   A. Hetero Phagosomes are secondary lysosomes formed by endocytosed phagosomes and primary lysosomes. Lysogenic digestion of endocytosed material is termed ***heterophagy***.
   B. Auto phagosomes are formed by fusion of cellular particles and primary lysosomes. The process of digestion of portion of a cell's own cytoplasmic constituents by its lysosomes is termed ***autophagy*.**
3. **Residual bodies:** Hetero Phagosomes and auto phagosomes after digestion and absorption are left with only residues and denatured enzymes within the vacuole, which are termed residual bodies.

Some important facts of Lysosomes:

1. Lysosomes serve as intracellular digestive system, so known as **digestive bags**.
2. They destroy any foreign material which enters the cell such as bacteria or virus.
3. Lysosomes also remove the worn out and poorly working cellular organelles by digesting them to make way for their new replacements. Since they remove cell debris, are known as **scavengers, cellular housekeepers or demolition squads.**
4. Lysosomes form a kind of **garbage disposal system** of cell
5. During breakdown of cell structure, when the cell gets damaged, lysosomes burst and the enzymes eat up their own cells. It is also known as suicide bags of a cell.

Function of Lysosomes: *It can be grouped together as functions of Lysosomes.*

A) CELLULAR DIGESTION: Lysosomal enzymes degrade proteins into dipeptides and carbohydrates onto monosaccharides. Sucrose & polysaccharides are not digested and remain in the lysosomal vacuoles.

B) AUTOPHAGY: By the process of autophagy lysosomes constantly remove cellular components like mitochondria etc. Cytoplasmic organelles become surrounded by smooth endoplasmic reticulum and lysosomes attach with it and discharge their contents into autophagic vacuole and the organelle is digested. Autophagy is a general property of eukaryotic cells.

C) DEVELOPMENTAL PROCESSES: Many developmental processes involve shedding or remodeling of tissues with removal of whole cells and extracellular material. It is observable in tadpole metamorphosis (regression of tail).

D) EXOCYTOSIS: Contents of the primary lysosome may be released into the medium by exocytosis and it occurs during replacement of cartilage by bone during development where osteoclasts release lysosomal enzymes. It can also occur in bone remodeling under influence of parathyroid hormone Crinophagy refers to the process by which secretory granules produced in excess are removed by lysosomes.

E) ENDOCYTOSIS: Lysosomes may fuse with vesicles or vacuoles formed by endocytosis and release their enzymes into it for digestion. The material for digestion may be food (protozoa) or a foreign body like parasite .The products of digestion are absorbed and assimilated leaving undigested which are released outside by exocytosis.

Hydrolytic Enzymes: Functions are as follows:

1. Acting on ester bonds, e.g. Phospholipase A1, phospholipase A2
2. Acting on glycosyl compounds, e.g. Lysozyme, neuraminidase etc
3. Acting on peptic bonds. e.g. Carboxyepetidase, dipeptidase etc
4. Acting on acid anhydrites ,e.g. Inorganic pyrophosphatase
5. Acting on other C-N bonds,e.g. amino acid napthylamidase
6. Acting on acid anhydrites ,e.g., inorganic pyrophosphates
7. Acting on P- N bonds, e.g. phosphoamidase
8. Acting on S--N bonds, e.g. Heparin sulfamidase

Ribosomes: Ribosomes are universal components of all living cells concerned with protein synthesis. In prokaryotic cells, they occur freely in the cytoplasm while in eukaryotic cells; occur freely in the cytoplasm or remain attached to the outer surface of the ER or nuclear membrane. Ribosomes are small, dense, granular, non membranous cell organelles made up of ribonucleoprotein (RNP) particles known as informosomes.

Structure of Ribosomes

1. Ribosomes are spherical or oval bodies.
2. Unlike other cytoplasmic organelles, they lack any membrane.
3. Each ribosome is made up of two unequal subunits that remain bound to each other by Mg2+ ions. The larger subunit is spherical and the smaller one is oval.
4. The two subunits join to form ribosomes of varying size like 70s and 80s (s stands for sedimentation coefficient or Svedberg units).
5. The subunits of prokaryotic 70s ribosomes are 30s and 50s while those of eukaryotic 80s ribosomes are 40s and 60s.
6. The 55s ribosomes are the smallest, containing 25s and 35s subunits and they are found in mitochondria and plastids of eukaryotic cells.
7. The RNA found in ribosomes is called ribosomal RNA (rRNA).

Functions of Ribosomes

1. Ribosome acts as a protein factory of a cell, site of protein synthesis within the cell.
2. Together with messenger RNA (mRNA), ribosomes form the template on which amino acids are joined to each other by peptide bonds to form the polypeptide chain or protein. In fact, it is the transfer RNA (tRNA) that brings the required amino acids to the ribosomes where they are assembled to form the new protein.
3. The GER (Golgi, ER) is extremely well developed in cells actively engaged in protein synthesis. The GER is characterized by the presence of attached ribosomes. These are present as polyribosomes or polysomes on the outer surface of GER; they are held together by mRNA and are often arranged in rosettes and spirals. The ribosomes are attached by their large 60s subunit.
4. The presence of special receptor sites for ribosomes is postulated. GER contains two transmembrane glycoproteins called ribophorin I and ribophorin II that are involved in mediating the attachment of ribosomes to the membrane.

Types of Microscopes

Light or compound microscope: The compound microscope is the oldest and still the most commonly used for studying the structure of organisms and cells. Light microscopy uses visible light and combination of lenses for magnification of images which are representatives of small samples. The microscopes optical in nature are old and are known to be designed in 1600. It is currently possible for capturing of image by light sensitive cameras in form of micrograph. As an alternate approach to light microscope techniques like scanning electron microscopy and transmission electron microscopy are used to get clear understanding of micro objects. Historically two basic configurations are known such as simple and compound with difference of number of lenses used. Currently compound microscopes are used which are known to be cheaper than digital microscopes.

Components of Light Microscope: Modern day microscopes are being designed to facilitate the viewing of samples by utilizing the transmitted light and contain the following components.

- ✓ Light Source like light or mirror
- ✓ Diaphragm and condenser lens
- ✓ Objective
- ✓ Ocular lens referred as eyepiece

Objective of light microscope is referred as cylinder with more than one lens and is made up of glass. The utility of objective in a light microscope is to collect the light from the sample and objective lenses can be of 4X, 5X, 10X, 20X, 40X, 50X, 60X, and 100X.

The objectives of the most microscopes are par focal where in lens differ from one microscope to the other. They are characterized on the basis of magnification and numerical aperture. In case of high magnification oil-immersion objectives are used which are designed specifically with proper utilization of refractive index matching oil which helps to get a good magnification of the image.

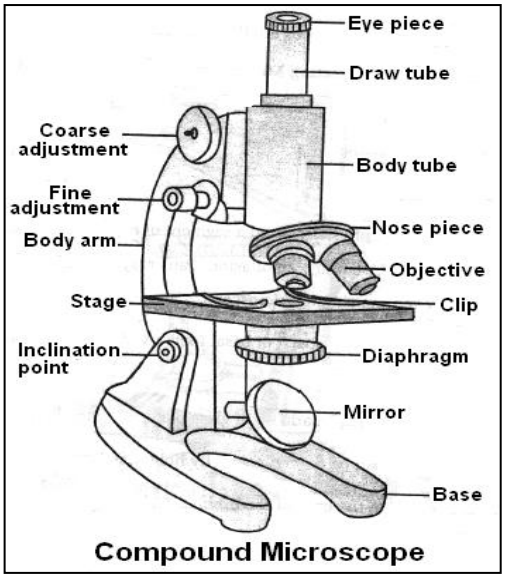
Compound Microscope

A. Lens Systems: are ***3*** types i.e., eyepiece, objectives & condenser.

1. **Eye piece (occular):** It lies at the top of the body tube. They are generally of 5x,6x,8x,10x,12x and 15x magnification. As per requirement they may be replaced by lower or higher magnification, it forms secondary image previously by the objective lens system.
2. **Objectives**: Objectives are attached with the nose piece. These contain lenses of different magnification. It is of 5x, 10x for low power and 40x, 4sx, 65x for higher power and 100x for oil immersion. It is above the object. It produces and magnifies the primary image of the specimen.
3. **Condenser**: It consists of condense lens system. It is beneath the specimen (objects) which is placed on the stage of microscope. It collects and focuses the light rays on the specimen.
4. **Diaphragm**: It is present below the stage. The diaphragmic knob controls the amount of light passes through the hole.
5. **Mirror**: Mirror is of concave and plane type. The concave mirror is used for the lower and higher power of magnification. The plane mirror is used for the dissection of object.

B. Mechanical parts: The mechanical-parts of the compound microscope are as follows.

1. **Draw tube**: It is hollow tube that holds eye piece at upper end and objectives at the lower end.
2. **Nose-piece**: It is revolving part for the adjustment of objectives. In the nose piece low and high power lenses are fitted.
3. **Coarse adjustment**: It is a big screw attached to the body tube on either side. It move body tube up and down and covers more distance in one rotation. It is used for low power adjustment.
4. **Fine adjustment**: It is a small screw attached to the arm on either side. It also moves the body tube up and down and covers small distance in one rotation. It is used for higher power, exact and sharp focusing of the object.

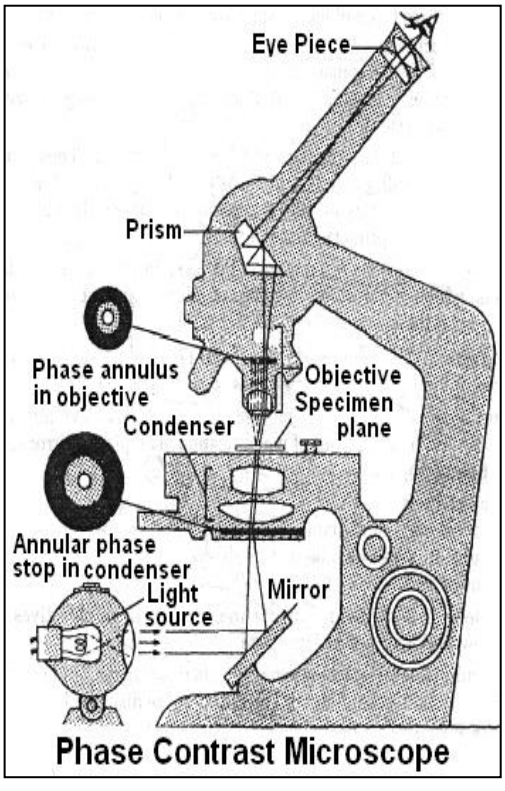


Compound Microscope

5. **Body arm (limb)**: It is stout part used to hold the microscope.
6. **Stage with Clips:** A rectangular part on which specimen is placed & fixed with clips.
7. **Inclination point:** It joins the lower (pillar) and upper parts of the microscope.
8. **Base (Foot)**: the base is lower part of microscope. It gives support to the microscope and bears its weight. It is horse shoe-shaped.

Phase contrast microscope: It was invented by Dutchman Fredrick Zernick in 1935. It is used to study living cells and tissues without staining. In this microscope the source of illumination is the ordinary visible light. Therefore its resolving power is the same as that of the light microscope.

1. It is a type of light microscopy that enhances contrast of transparent and colourless object by influencing the optical path of light.
2. It is able to show component in cell or bacteria.
3. Phase-contrast microscopy used to measure the refractive index and solid concentration of cell structure by immersion refractometry method.
4. This method involves the immersion of cells is an isotonic solution, the refractive index can be varied in small increments
5. The optical system of the phase contrast microscope differ from ordinary optical microscope only in addition of:
   a. A sub-stage annular diaphragm to illuminate object with narrow cone of light.
   b. A diffraction plate mounted in the objective.
6. The phase change Ø or optical path difference begin by an object is expressed as, $Ø = (n_o - n_m)$ t ------ where n_o and n_m are the refractive index of the object and t -is the thickness of object.



Phase Contrast Microscope

7. This equation show that if the refractive index of the mounting medium is made equal to that of the objects, the phase change will be zero-and object will be invisible or minimum contrast when viewed with the phase-contrast microscope.
8. When the refractive index of the mounting medium is greater or less than that of object, the object will appear in dark or bright contrast respectively.
9. Diffraction plate is used in the phase-contrast microscope to separate diffracted light, so that they can be altered in phase relative, to each other to give increased or decreased brightness of the object.
10. Phase-contrast microscope is a vital instrument in biological and medical research.
11. Study of cell division, cell movements and membrane formation can easily recorded by phase-contrast microscope.

Electron microscope:

It was invented by Knoll and Buska in 1931. It has electromagnetic lenses which are coils of wire enclosed in a soft iron case.

Its essential parts are: Metal (Tungsten) Filament, Electromagnetic condenser lens, electromagnetic objective lens, electromagnetic projector lens.

Living cells and tissues can not be studied under light and electron microscope. These are killed, fixed in certain chemical solutions, sectioned and stained to provide contrast.

Electron Microscopes are scientific instruments that use a beam of highly energetic electrons rather than a light beam to examine objects on a very fine scale. It can get a magnification upto 2 million times. It resolves fine structure of the cell. The resolving power of electron microscope is 0.1 nm. Electron microscopy is a powerful tool for the study of macromolecules and cellular ultra structure.

Electron microscope provides tremendous magnification, as a result of the higher resolution which is obtained by the extremely short wavelength of electron beams which are used to magnify the specimens. An electron microscope uses a beam of electrons and magnetic field in contrast to a beam of light used in optical microscope. The resolving power of electron microscope theoretically is around X400, 000 times with respect to the wavelength of electrons used that range approximately 0.005nm which corresponds to 0.05Ao where one Angstrom equals 10-8cm.

Structure and Function of Electron Microscope: The electron microscope consists of an electron source, an anode, magnetic lenses, apertures, specimen stage and image recording system all operates in a high vacuum.

Electron microscope consists of cylindrical tube of about 2 meters long which is completely devoid of air. Electrons microscope visualizes using thin beam of rapidly moving electrons that interferes with specimen placed in the tube.

Electrons are emitted by the cathode at the top of the tube and then accelerate by the anode. They then pass through the small aperture that forms them into a beam and into the vacuum inside the tube.

The part of the microscope that generates the electron beam is sometimes called the electron gun. The beam is maintained along the tubes by means of electromagnetic lenses. These are coils that surround the tube at given intervals. The electromagnetic field emitted by the coils focuses the beam at the centre of the tube.

Electrons encounter the specimen are either absorbed, scattered or pass through it. Because different regions of the specimen are variously transparent to electrons different amounts of electrons with changed energy pass through these regions. At the end of the tube the electrons are collected on florescent photographic film or on screen that generate the energy of the specimen.

The beam that reaches the film consists of different amount of electrons that pass through particular region of the specimen. This difference is responsible for the contrast in the film.

The two types of electron microscopy are discussed below:

I) Transmission electron microscopy:

Specimens to be examined on electron microscope is prepared as a very thin dry film on small screens and is introduced inside between the magnetic condenser and the magnetic objective, thus enabling the magnified image to be examined on a fluorescent screen or is recorded on a photographic plate by a camera that is built into the instrument.

II) Scanning electron microscopy:

Specimen to be examined is subjected to a narrow electron beam which rapidly scans the surface of the specimen, causing of a release of shower of secondary electrons; the intensity of which is collected by a detector generating electronic signals which are then scanned in a television to produce the image.

Limitations of Electron Microscope

Since the specimens observed are under very high vacuum, cells in living state cannot be examined and the drying process involved in the preparation of specimens may alter few morphological characteristics. Since electron beams have very low penetration power, very thin sections are required.

Advantages of Electron Microscope

- ✓ Higher magnification and lens power can be increased by adjusting currents.
- ✓ High resolving power.
- ✓ Powerful tool to study ultrastructure.

Disadvantages of electron microscope-

- ✓ Need to kill and ‘fix’ cells
- ✓ Difficult and time consuming
- ✓ Expensive (approx £200,000)

Cytology of Cancer: Cancer is a proliferation of cells which grow in an uncontrolled manner, invading local tissues and spreading widely through the blood or lymphatics to produce secondary deposits, or metastases in distant parts of the body.

During normal development and growth the cells in our body divide mitotically and get differentiated to specialized cells of the tissues. The processes of cell mitosis, growth and differentiation are controlled by cellular genes. Cancer is caused due to mutation or abnormal activation of such genes. such a mutation can happen in a single cell. Thus it may be **monoclonal** in origin. With further growth of cancer, additional mutations may occur in the daughter cells giving rise to subclones. The mutated cells may remain as heterogeneous cancer cells. Among these subclones some may have greater capacity and metastasize to distant tissues. They may also remain more resistant to damage from various anticancer treatments.

The cancer cells have characteristic properties. They can be differentiated from normal cells under microscopic observation. These cells have large nuclei. In each cancer cell, the ratio of nucleus to cytoplasm is high. They have prominent nucleoli. The cells can grow indefinitely in culture medium. As component cell of a tissue they remain less differentiated. Even after getting organized into tissues unlike other cells they do not lose replicative capacity. Cancer cells have the ability to invade surrounding tissues.

The sequence of events that convert a normal cell into a cancer cell is called **carcinogenesis**. The process of carcinogenesis includes, intiation, growth, promotion, **conversion**, propagation and progression. Progression includes the processes of **invasion** and **metastasis**.

Mature cancers have relatively uncontrolled growth, behaviour. As other normal cells they do not show any of the normal intracellular and extracellular growth control mechanisms. Initially the cancer cells have an exponential growth. Gradually their growth surpasses blood vascular supply. This results in slowing down of growth.

Types of Cancer: Cancers are classified on the basis of their location/the body site from which they arise.

1. Carcinomas: These cancers are located in the epithelial lining of internal organs or glands. For e.g., Breast cancer, Stomach cancer etc., Cancers of epithelial glands are called Adenoma. About 85% of all tumors are of this kind.

2. Melanomas: These are cancerous growths of melanocytes (skin cells).

3. Sarcomas: These cancers are located in connective and muscular tissues. These are derived from tissues of mesodermal origin that is bone, fat and cartilage. If cancer affects the Lymphatic system, it is called Lymphomas. For e.g., Hodgkin's disease in man is characterized by enlargement of the lymph nodes, spleen etc. Lipomas affect the adipose tissue and osteoma affects the bones.

4. Leukemia (Blood cancer): These are characterized by increased WBC count of the blood (up to 2, 00,000 - 1000, 000/mm3) due to their increased formation in the bone marrow. This decreases the erythropoiesis and RBC count. These are most common types of cancer affecting the children below 15 yrs of age. Bone marrow transplantation is recommended.

5. Myeloma: These are malignant tumors occurring in middle aged and older people that interfere with the blood - cell producing function of bone marrow and cause anemia.

Microtechnique:

Microtechnique : Object to be studied under a microscope needs preparation. If the object is small enough through which light can pass and the study in finest details is not required a whole mount preparation will be sufficient: In case of large objects they are to be cut in thin sections for the study. This is called histological preparations.

Collection of Tissue: For sectioning, tissues are to be collected from live specimens. In small vertebrates, the animal is paralyzed by damaging the brain. In large vertebrates, the same result is achieved by striking the head against a hard object. In still large species the tissues are collected immediately after killing the animal for some other purpose or a small piece of tissue is cut by surgical operation

Fixation: Killing the tissue without any structural or chemical change is known as fixing. The tissues are cut into small pieces, washed with physiological solution, if required, immersed in the fixative and left these for a definite period.

The common fixatives used are bouins fluid, Zenker's fluid and Carnoy fluid etc. The specimen tubes with the fixative and tissues should be subjected to a vacuum pump for a few minutes to remove the air in the tissue which may enter during collection.

Fix the tissue in Bouin's fluid or any suitable fixatives for 24 hours. Wash the tissue with lithium carbonate solution for 3 -4 times. Keep the tissue in 70%, 90%, 100% alcohol for 24 hours in each. Take 50:50 cedarwood oil and absolute alcohol. Then keep the tissue in cedarwood oil for 24 hours. Pass it to benzene and give 3 -4 washes. Remove benzene completely and pass it to pure wax. Keep at 60^{o}C for half an hour. Change the wax for 3 -4 times then the tissue is ready for block preparation.

Dehydration: Dehydration means removal of water from the tissues. It prevents decaying and maintains shape and size of tissues or cells. Dehydration is done by passing the mounting material through various grades of alcohols, such as 30,50,70,90 and 100% alcohols. The dehydration is carried out in corked or glass stoppered tubes.

Block Preparations: Make blocks either in metal L-shaped or in cavity blocks. Take two metal blocks, are place opposite to each other to prepare a square of it. First apply little amount of glycerin on their internal surface of block. Pour hot melted wax in the L blocks. Then add tissue and more melted wax to fully cover. Keep the block for two to three hours tiIl the melted wax, becomes cool and solid. Remove the metal L block and a square wax block is ready for microtomy.

Microtomy: The blocks are cut by rotatory microtome. Trim the block properly by cutter and make square of that block. Fix the block to the holder. The block holder is then fixed to the micro-tome; Microtome is adjusted to 5-6µ thickness. Ribbon must be cut in low temperatures only (30^{o}C). Keep clean slides ready. Apply egg albumen over the slide and rub it by last finger. The egg albumen helps in sticking the sections over glass surface. Ribbons should be kept upto more than half of the slide. Space should be left for putting labels over the slide. Flatten the section over a hot plate. The temperature of the hot plate should be nearly 35-40 ^{o}C. Add a few drops of water below the ribbons. As the water is heated, ribbons become expanded by semi-melting of the wax. After all the section become flattened, drain off water and leave the slide over night for drying the ribbons.

Staining: Sections of tissue are usually stained with two dyes to bring contrast between different histological structures. Double staining practiced in class work is with haematoxylin and eosin. The haematoxylin is basic dye imparts blue colour to acidic materials, viz. nucleic acid, which are concentrated in the nucleus and only scattered in the cytoplasm. The eosin is an acid dye and the cytoplasmic material being basic in nature is stained by it. The result is the nucleus and only a small fraction of the cytoplasm appears blue, while the rest of the cell takes up red colour.

Techniques

1. The slide with the paraffin ribbon containing sections of tissue is dipped in xylol for about 5 minutes.
2. The paraffin wax dissolves.
3. Slide is transferred to absolute alcohol.
4. Time in each grade of alcohol is about 5 minutes.
5. Slide is kept in haemotoxylin stain for about 10 -15 minutes.
6. Time taken for eosin stain is about 10 minutes.
7. Put a small amount of Canada balsam or DPX on the slide.
8. Put the coverslip slowly.
9. If air bubbles are locked between the coverslip and the slide, it may be removed by leaving the slide over night on a hot plate.
10. A good stained slide reveals pinkish colour of cytoplasm and blue colour of nuclei.

Staining Procedure

Slide

Xylene (Xylol)	5 minutes
100% alcohol	5 minutes
90% alcohol	5 minutes
70% alcohol	5 minutes
50% alcohol	5 minutes
30% alcohol	5 minutes
Distilled water	One to two dip.
Haematoxylin	10-15 minute
30% alcohol	5 minute
50% alcohol	5 minutes
7O% alcohol	5 minutes
Eosin stain	10 minutes
90% alcohol	5 minutes
100% alcohol	5 minutes
Xylene	5 minutes

Mount in Canada balsam or DPX.

Preparation of stain

I. Haematoxylin

a) Haematoxylin 1gm
b) Absolute alcohol 10 ml
c) Potassium alum 20 gms
or Ammonium alum

d) Distilled water 200 ml
e) Mercuric oxide 0.5gm
f) Glacial acetic acid 8ml

Dissolve the haematoxylin in absolute alcohol and potassium alum is dissolved in hot distilled water. Heat the mixture to boiling point and add the mercuric oxide. Cool rapidly and filter the stain. It is ready for use.

II. Eosin

a) Eosin 1gm
b) Distilled water 20ml
c) Alcohol (95%) 80 ml
d) Glacial acetic acid 0.5 ml

Preparation of Fixatives

Bouin's Fluid: Dissolve I -10 gm of picric acid in 100 ml of distilled water. After 48 hours the solution is ready for Use.

Definition of Genetics: Genetics, a discipline of biology, is the science of genes, heredity, and variation in living organisms. Genetics deals with the molecular structure and function of genes, gene behavior in context of a cell or organism (e.g. dominance and epigenetics), patterns of inheritance from parent to offspring, and gene distribution, variation and change in populations, such as through Genome-Wide Association Studies. Given that genes are universal to living organisms, genetics can be applied to the study of all living systems, from viruses and bacteria, through plants and domestic animals, to humans.

The fact that living things inherit traits from their parents has been used since prehistoric times to improve crop plants and animals through selective breeding. However, the modern science of genetics, which attempts to understand the process of inheritance, only began with the work of Gregor Mendel in the mid 19th century. Although he did not know the physical basis for heredity, Mendel observed that organisms inherit traits by way of discrete units of inheritance, which are now called genes.

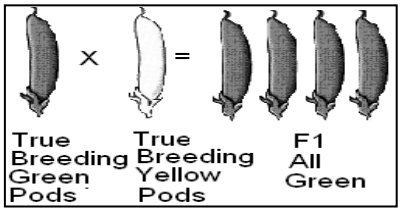
Although genetics plays a large role in the appearance and behavior of organisms, it is the combination of genetics with what an organism experiences that determines the ultimate outcome. For example, while genes play a role in determining an organism's size, the nutrition and health it experiences after inception also have a large effect.

Genotype and Phenotype: From Mendel's law of segregation, the alleles for a trait separate when gametes are formed. These allele pairs are then randomly united at fertilization. If pair of alleles for a trait is same they are called homozygous. If they are different they are called heterozygous. In the first example (Figure A), the F1 plants were all heterozygous for the pod color trait. Their genetic makeup or genotype was (Gg). Their phenotype or expressed physical trait was green pod color.

The F2 generation pea plants (Figure B) showed two different phenotypes (green or yellow) and three different genotypes (GG, Gg, or gg). The genotype determines the phenotype that is expressed. The F2 plants that had a genotype of either (GG) or (Gg) were green. The F2 plants that had a genotype of (gg) were yellow.

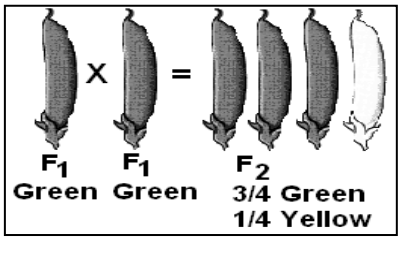
The phenotypic ratio that Mendel observed was 3:1, 3/4 green plants to 1/4 yellow plants. The genotypic ratio however was 1:2:1. The genotypes for the F2 plants were 1/4 homozygous (GG), 2/4 heterozygous (Gg), and 1/4 homozygous (gg).

Mendel's Law of Segregation: When Mendel performed cross-pollination between a true-breeding yellow pod plant and a true-breeding green pod plant, he noticed that all of the resulting offspring, F1 generation, were green.

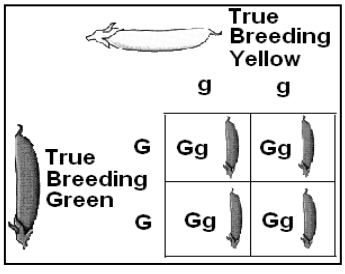


Mendel's Law of Segregation: He then allowed all of the green F1 plants to self-pollinate. He referred to these offspring as the F2 generation. Mendel noticed a 3:1 ratio in pod color.

About 3/4 of the F2 plants had green pods and about 1/4 had yellow pods. From these experiments Mendel formulated, is now known as Mendel's law of segregation.



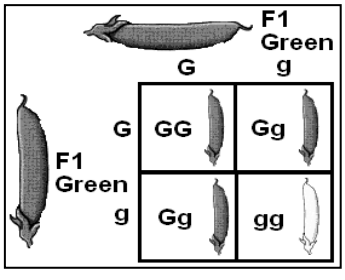
Mendel's Law of Segregation: Mendel's law of segregation states that allele pairs separate or segregate during gamete formation, and randomly unite at fertilization. There are four main concepts involved in this idea. They are:



1. There are alternative forms for genes. This means that a gene can exist in more than one form. For example, the gene that determines pod color can either be (G) for green pod color or (g) for yellow pod color.

2. For each characteristic or trait organisms inherit two alternative forms of that gene, one from each parent. These alternative forms of a gene are called alleles. The F1 plants in Mendel's experiment each received one allele from the green pod parent plant and one allele from the yellow pod parent plant. True-breeding green pod plants have (GG) alleles for pod color, true-breeding yellow pod plants have (gg) alleles, and the resulting F1 plants have (Gg) alleles.

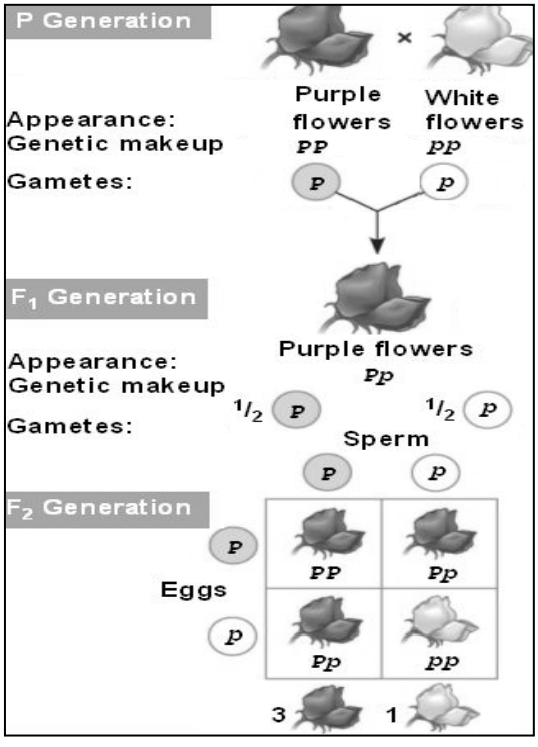
Mendel's Law of Segregation:



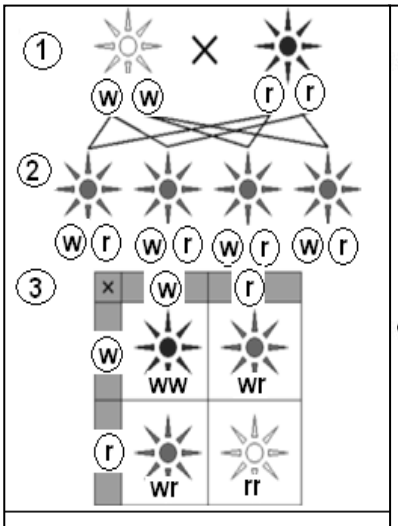
1. **When gametes (sex cells) are produced, allele pairs separate or segregate leaving them with a single allele for each trait**. This means that sex cells contain only half the compliment of genes. When gametes join during fertilization the resulting offspring contain two sets of alleles, one allele from each parent. For example, the sex cell for the green pod plant had a single (G) allele and the sex cell for the yellow pod plant had a single (g) allele. After fertilization the resulting F1 plants had two alleles (Gg).
2. **When the two alleles of a pair are different, one is dominant and the other is recessive**. This means that one trait is expressed or shown, while the other is hidden. For example, the F1 plants (Gg) were all green because the allele for green pod color (G) was dominant over the allele for yellow pod color (g). When the F1 plants were allowed to self-pollinate, 1/4 of the F2 generation plant pods were yellow. This trait had been masked because it is recessive. The alleles for green pod color are (GG) and (Gg). The alleles for yellow pod color are (gg).

Mendel's Laws: Mendel discovered that when crossing white flower and purple flower plants, the result is a mix of the two, the offspring was purple flowered. He then conceived the idea of heredity units, which he called "factors", one of which is a recessive characteristic and the other dominant. Mendel said that factors, later called genes, normally occur in pairs in ordinary body cells, yet segregate during the formation of sex cells. Each member of the pair becomes part of the separate sex cell. The dominant gene, such as the purple flower in Mendel's plants, will hide the recessive gene, the white flower. After Mendel self-fertilized the F1 generation and obtained the 3:1 ratio, he correctly theorized that genes can be paired in three different ways for each trait: PP, pp, and Pp. The capital "P" represents the dominant factor and lowercase "p" represents the recessive. Mendel stated that each individual has two factors for each trait, one from each parent. The two factors may or may not contain the same information. If the two factors are identical, the individual is called homozygous for the trait. If the two factors have different information, the individual is called heterozygous. The alternative forms of a factor are called alleles.

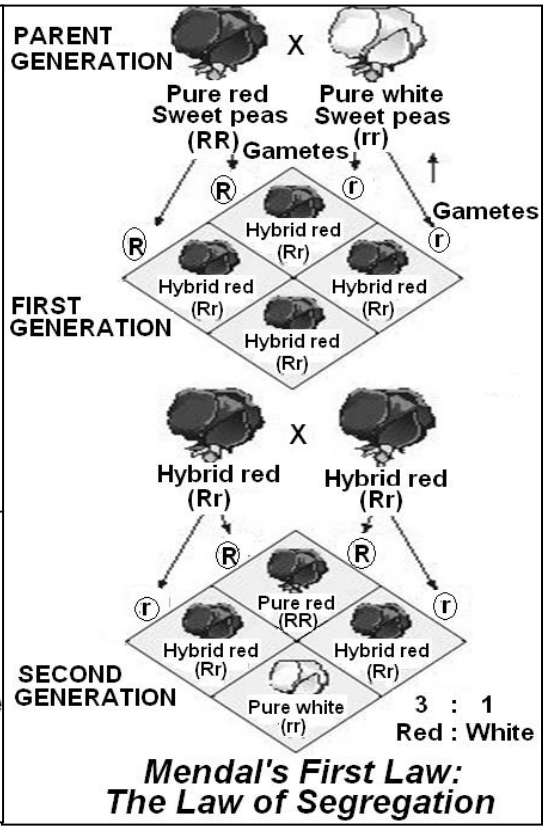
The genotype of an individual is made up of the many alleles it possesses. An individual's physical appearance, or phenotype, is determined by its alleles as well as by its environment. An individual possesses two alleles for each trait; one allele is given by the female parent and the other by the male parent. They are passed on when an individual matures and produces gametes: egg and sperm. When gametes form, the paired alleles separate randomly so that each gamete receives a copy of one of the two alleles. The presence of an allele doesn't promise that the trait will be expressed in the individual that possesses it. In heterozygous individuals the only allele that is expressed is the dominant. The recessive allele is present but its expression is hidden. Mendel summarized findings in two laws; the **Law of Segregation** and the **Law of Independent Assortment**.



Law of Segregation (The "1st Law"): The Law of Segregation states that when any individual produces gametes, the copies of a gene separate so that each gamete receives only one copy. A gamete will receive one allele or the other. In meiosis the paternal and maternal chromosomes get separated and the alleles with the traits of a character are segregated into two different gametes.



The colour alleles of Mirabilis jalapa are not dominant or recessive. (P) Parental generation (F1) F1 generation (F2) F2 generation. the "red" and "white" allele together make a "pink" phenotype, reulting in a 1:2:1 ratio of red:pink:white in the F2 generation.



Law of Independent Assortment (The "Second Law")

The Law of Independent Assortment, also known as "Inheritance Law" states that alleles of different genes assort independently of one another during gamete formation. While Mendel's experiments with mixing one trait always resulted in a 3:1 ratio between dominant and recessive phenotypes, his experiments with mixing two traits (dihybrid cross) showed 9:3:3:1 ratios. But the 9:3:3:1 table shows that each of the two genes is independently inherited with a 3:1 phenotypic ratio.

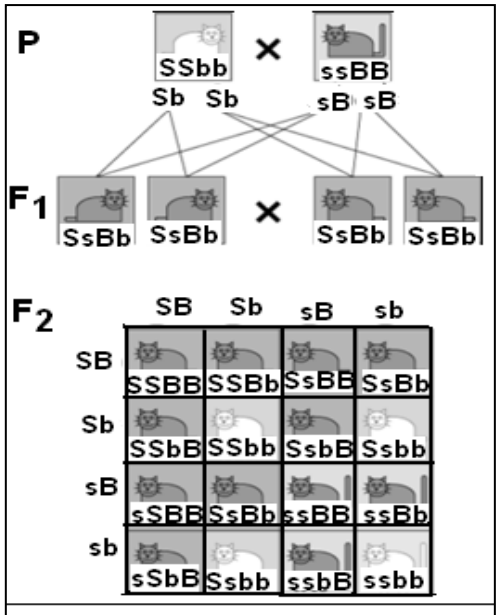


Figure 2: The phenotypes of two independent traits show a 9:3:3:1 ratio in the F2 generation. In this example, coat color is indicated by B (brown, dominant) or b (white) while tail length is indicated by S (short, dominant) Or s (long). When parents are homozygous for each trait ('SSbb and ssBB), their children in the F1 generation are heterozygous at both loci and only show the dominant phenotypes. If the children mate with each other, in the F2 generation all combination of coat color and tail length occur: 9 are brown/short (purple boxes), 3 are white/short (pink boxes), 3 are brown/long (blue boxes) and 1 is white/long (green box).

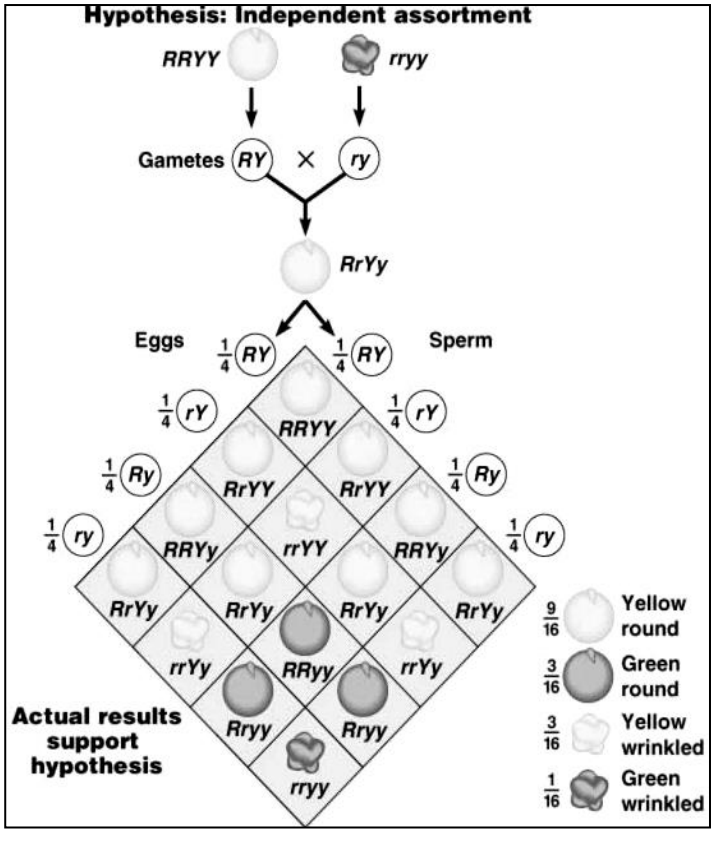
Mendel concluded that different traits are inherited independently of each other, so that there is no relation, for example, between a cat's color and tail length. This is actually only true for genes that are not linked to each other. During meiosis I in eukaryotic organisms, specifically metaphase I of *meiosis*, to produce a gamete with a mixture of the organism's maternal and paternal chromosomes. Along with chromosomal crossover, this process aids in increasing genetic diversity by producing novel genetic combinations.

Of the 46 chromosomes in a normal diploid human cell, half are maternally-derived (from the mother's egg) and half are paternally-derived (from the father's sperm). This occurs as sexual reproduction involves the fusion of two haploid gametes (the egg and sperm) to produce a new organism having the full complement of chromosomes.

During gametogenesis—the production of new gametes by an adult—the normal complement of 46 chromosomes needs to be halved to 23 to ensure that the resulting haploid gamete can join with another gamete to produce a diploid organism. An error in the number of chromosomes, such as those caused by a diploid gamete joining with a haploid gamete, is termed aneuploidy.

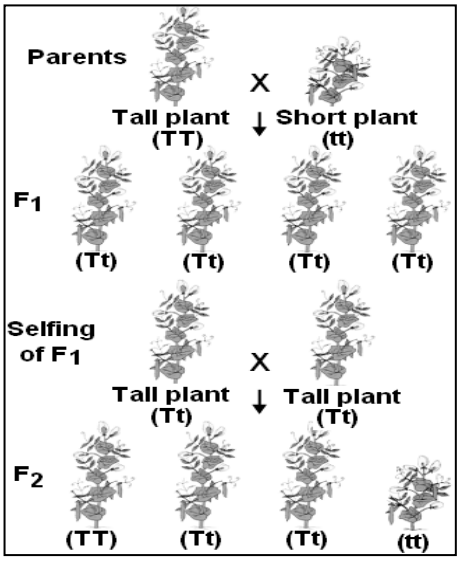
In independent assortment the chromosomes that end up in a newly-formed gamete are randomly sorted from all possible combinations of maternal and paternal chromosomes. Because gametes end up with a random mix instead of a pre-defined "set" from either parent, gametes are therefore considered assorted independently. As such, the gamete can end up with any combination of paternal or maternal chromosomes. Any of the possible combinations of gametes formed from maternal and paternal chromosomes will occur with equal frequency.

Mechanism of Independent Assortment: can be understood easily by assuming that the homozygous pea plant with yellow round seeds has the alleles YY and RR for yellow colour and roundness of the seed, respectively. Similarly the homozygous peas plant with green wrinkled seeds contain the alleles yy and rr for the green colour and wrinkledness of seeds. The gametes which are produced by YYRR and yyrr plants are YR and yr types respectively. When both parents are crossed the union of both types of gametes takes place to give F1 hybrid (YyRr). The F1 hybrid have been found to contain yellow round seeds showing the dominance of allele Y for yellow colour over the recessive allele y for green colour and the dominance of allele R for roundness over the recessive allele for wrinkledness of seed. Now F1 hybrids have four types of allele viz. Y for yellow colour, y for green colour, R for round shape and r for wrinkledness of seed. Thus four types of alleles are assorted independently to produce four types of gametes. Viz., YR, Yr, yR, and yr. Thus the sixteen F2 individuals have the ratio of 9 yellow round : 3 yellow wrinkled : 3 green round and 1 green wrinkled.



Example: When Mendel cross pollinated two plants with contrasting characters he observed the following

- After hybridization, the F_1 generation obtained resembled only one of its parents (all tall; no dwarf).
- When two plants from F_1 generation were self pollinated, the second filial progeny or F_2 generation was obtained.
- Revival of the trait that was unexpressed in F_1 (dwarf/short) was observed in some F_2 progeny. Both traits, tall and dwarf/short, were expressed in F_2 generation in ratio 3:1.
- Mendel proposed that something is being passed unchanged from generation to generation (presently called genes).
- Factors contain and carry hereditary information.
- Traits may not show up in an individual but are passed on to the next generation.



Gene Interaction – Definition: Most of the characters of living organisms are controlled / influenced / governed by a collaboration of several different genes. This condition where a single character is governed by two or more genes and every gene affect the expression of other genes involved (these genes affect each other's expression) is known as gene interaction.

Gene interactions may involve two or more pairs of genes. But all the gene interactions have the two pairs of non-allelic genes, affecting the phenotypic expression of same character. These interactions produce modified dihybrid ratios.

Types of Gene Interactions

- ✓ Allelic gene interaction
- ✓ Non-allelic gene interaction

Non-allelic gene interaction

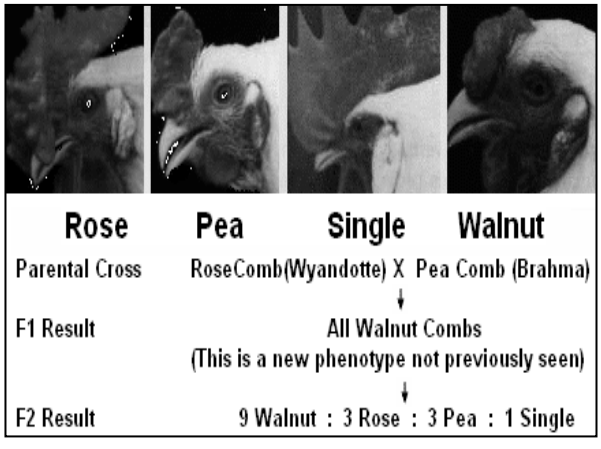
Expression of character is produced by interaction between two or more genes. The interactions listed, as inter-allelic and intra-allelic type.

Inter-allelic

- ✓ Without modification of normal F2 ratio
- ✓ With modification of normal F2 ratio

Chicken	Phenotype
Wyandotte	Rose Comb
Brahmas	Pea Comb
Leghorns	Single Comb

Such kinds of interactions modify the normal F_2 ratio (9:3:3:1). Various types of such interactions are as below. The genes of an individual do not operate isolated from one another, but obviously are functioning in a common cellular environment. Thus, it is expected interactions between genes would occur. Bateson and Punnett performed a classical experiment that demonstrated genetic interactions. They analyzed the three comb types of chicken known to exist at that time:



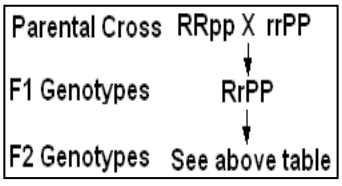
Result: The F1 differed from both parents and two new phenotypes not seen in the parents appeared in the F2. How can this result be explained? The first clue is the F2 ratio. We have seen this ratio before when the F1 from a dihybrid cross is selfed (or intermated). This observation suggests that two genes may control the phenotype of the comb. The gene interactions and genotypes were determined by performing the appropriate testcrosses. A series of experiments demonstrated that the genotypes controlling the various comb phenotypes are as follows. It was shown that the genotypes of initial parents were:

Phenotypes	Genotypes	Frequency
Walnut	R_P_	9/16
Rose	R_pp	3/16
Pea	rrP_	3/16
Single	Rrpp	1/16

Rose = *RRpp*

Pea = *rrPP*

Therefore, genotypically the cross was: The development of any individual is obviously the expression of all the genes that are a part of its genetic makeup. Therefore, it is not an unexpected conclusion that more than one gene could be responsible for the expression of a single phenotype.



Modification of Dihybrid Phenotypic Ratio: The classical or Mendelian phenotypic ratio of 9:3:3:1 is obtained only when the alleles at both gene loci display dominant and recessive relationships. If one or both gene loci have incompletely dominant alleles, or codominant alleles or lethal alleles, the dihybrid ratio becomes modified variously, such as: ***3: 6 : 3 : 1 : 2 : 1 ratio***

When the dihybrid parents have dominant and recessive alleles at one gene Iocus and codominant alleles at second gene locus, the F2 ***9 : 3 : 3 : 1.***

Phenotypic ratio becomes ***3 : 6 : 3 : 1 : 2 : 1.***

Example: In cattles. Hornless or polled (P) condition is dominant to horned (p) condition, and white (W) coat colour is codominant to red coat colour (w). The mating of homozygous white, polled (hornless) cattle (PP WW) with homozygous red horned cattle (pp ww) will produce F1 heterozygotes with the phenotype of hornless or polled, roan and genotype of Pp Ww. These hornless or polled roan F1 heterozygotes produce a F2 progeny in the ratio of ***3 : 6 : 3 : 1 : 2 : 1:*** The ratio of ***3: 6: 3: 1: 2: 1*** also can be get by cross-multiplying the F2 monohybrid phenotypic ratios of dominant recessive alleles ***(3: 1)*** and codominant alleles ***(l : 2 : 1), i.e., (3 : 1)X(1 : 2 : 1) =3 : 6 : 3 : 1 : 2 : 1.***

F_2 analysis of figure:

Phenotype	**Hornless (polled)white**	**Hornless (polled) roan**	**Hornless (polled) red**	**Horned white**	**Horned roan**	**Horned red**
Genotype	PP WW Pp WW pP WW	PP Ww PP wW Pp Ww Pp wW pP Ww pP wW	PP ww Pp ww pP ww	pp WW	Pp Ww Pp wW	pp ww
Ratio	3	6	3	1	2	1

1: 2: 1: 2: 4: 2: 1: 2: 1 ratio

When each parent of a dihybrid cross has incompletely dominant alleles at both gene loci, then in F2 large numbers of phenotypic classes are produced. Example: In snapdragons, red flower colour (R) is incompletely dominant to white flower colour (r) and broadness of leaf (B) is incompletely dominant to narrowness of leaf (b). The dihybrid cross between red, broad plant (RR BB) and white, narrow plant (rr bb) produces F1 heterozygotes having pink flowers and leaves of intermediate width and genotype of Rr Bb. These F1 heterozygotes produced the F2progeny in 9 phenotypic classes as shown in Figure. The phenotypic ratio of ***1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1*** can also be obtained by cross multiplying the F2 monohybrid ratios of each gene (R or B),

i.e., (1 : 2 : 1) X (1 : 2 : 1) = 1: 2 : I : 2 : 4 : 2 : 1:2:1. 3: 1: 6: 2 ratios

Epistasis: Epistasis is the phenomenon where the effects of one gene are modified by one or several other genes, which are sometimes called modifier genes. The gene whose phenotype is expressed is said to be epistatic, while the phenotype altered or suppressed is said to be hypostatic. Epistasis can be contrasted with dominance, which is an interaction between alleles at the same gene locus. Epistasis is often studied in relation to Quantitative Trait Loci (QTL) and polygenic inheritance. In general, the fitness increment of any one allele depends in a complicated way on many other alleles; each gene was considered to make its own characteristic contribution to fitness, against an average background of other genes.

Epistasis and genetic interaction refer to different aspects of the same phenomenon. The term epistasis is widely used in population genetics and refers especially to the statistical properties of the phenomenon, and does not necessarily imply biochemical interaction between gene products. However, in general epistasis is used to denote the departure from 'independence' of the effects of different genetic loci. Confusion arises due to the varied interpretation of 'independence' between different branches of biology.

Examples of tightly linked genes having epistatic effects on fitness are found in supergenes and the human major histocompatibility complex genes. The effect can occur directly at the genomic level, where one gene could code for a protein preventing transcription of the other gene. Instead, the effect can occur at the phenotypic level.

	AB	***Ab***	***aB***	***Ab***
No epistasis (additive across loci)	2	1	1	0
Synergistic epistasis	3	1	1	0
Antagonistic epistasis	1	1	1	0

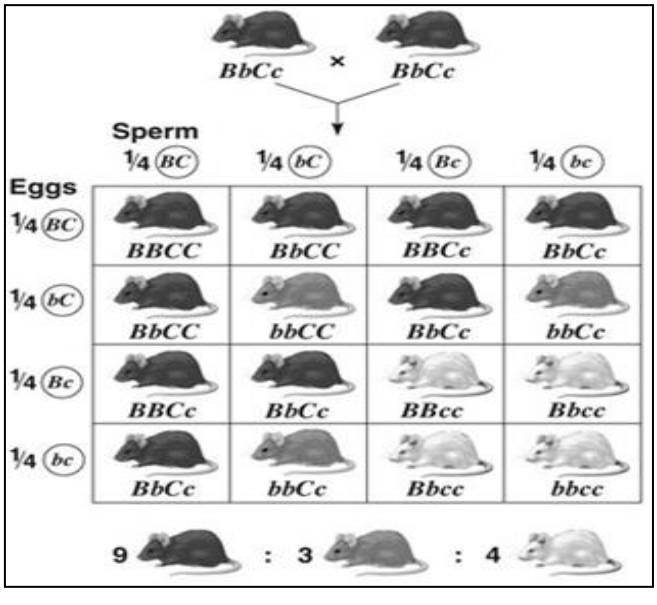
For example, the gene causing albinism would hide the gene controlling color of a person's hair. In another example, a gene coding for a widow's peak would be hidden by a gene causing baldness. Fitness epistasis is one cause of linkage disequilibrium.

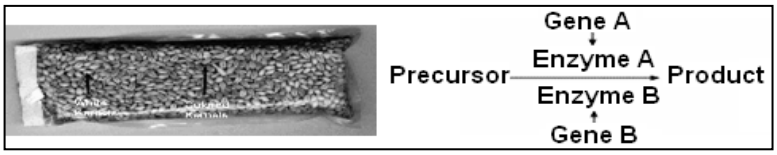
Two-locus epistatic interactions can be either synergistic (enhancing the effectiveness) or antagonistic (reducing the activity).

Hence, we classify as	
Trait values	**Type of epistasis**
AB = Ab + aB − ab	No epistasis, additive inheritance
AB > Ab + aB − ab	Synergistic epistasis
AB < Ab + aB − ab	Antagonistic epistasis

In the example of a haploid organism with genotypes (at two loci) AB, Ab, aB or ab, we can think of the following trait values where higher values suggest greater expression of the characteristic (the exact values are simply given as examples):

The interactions of the two genes which control comb type was revealed because we could identify and recognize the 9:3:3:1. Other genetic interactions were identified because the results of crossing two dihybrids produced a modified Mendelian ratio. All of the results are modifications of the 9:3:3:1 ratio.





Example 1: 15:1 Ratio **Phenotypes:** Kernel Color in Wheat: For this type of pathway a functional enzyme A or B can produce a product from a common precursor. The product gives color to the wheat kernel. Therefore, only one dominant allele at either of the two loci is required to generate the product. Thus, if a pure line wheat plant with a colored kernel (genotype = AABB) is crossed to plant with white kernels (genotype = aabb) and the resulting F1 plants are selfed, a modification of the dihybrid 9:3:3:1 ratio will be produced. The following table provides a biochemical explanation for the 15:1 ratio.

Genotype	Kernel Phenotype	Enzymatic Activities
9 A_B_	colored kernels	functional enzymes from both genes
3 A_bb	colored kernels	functional enzyme from the A gene pair
3 aaB_	colored kernels	functional enzyme from the B gene pair
1 aabb	colorless kernels	non-functional enzymes produced at both genes

If we sum the three different genotypes that will produce a colored kernel we can see that we can achieve a 15:1 ratio. Because either of the genes can provide the wild type phenotype, this interaction is called duplicate gene action.

Example 2: 9:7 Ratios

Example: Flower color in sweet pea. If two genes are involved in a specific pathway and functional products from both are required for expression, then one recessive allelic pair at either allelic pair would result in the mutant phenotype. Graphically shown in the diagram.

If a pure line pea plant with colored flowers (genotype = CCPP) is crossed to pure line, homozygous recessive plant with white flowers, the F1 plant will have colored flowers and a CcPp genotype. The normal ratio from selfing dihybrid is 9:3:3:1, but epistatic interactions of the C and P genes will give a modified 9:7 ratio. The following table describes the interactions for each genotype and how the ratio occurs.

Genotype	Flower Color	Enzyme Activities/TH>
9 C_P_	**Flowers colored; anthocyanin produced**	**Functional enzymes from both genes**
3 C_pp	**Flowers white; no anthocyanin produced**	**p enzyme non-functional**
3 ccP_	**Flowers white; no anthocyanin produced**	**c enzyme non-functional**
1 ccpp	**Flowers white; no anthocyanin produced**	**c and p enzymes non-functional**

Because both genes are required for the correct phenotype, this epistatic interaction is called complementary gene action.

Example 3: 12:3:1 Ratio

Phenotype: Fruit Color in Squash: With this interaction, color is recessive to no color at one allelic pair. This recessive allele must be expressed before the specific color allele at a second locus is expressed. At the first gene white colored squash is dominant to colored squash, and the gene symbols are W=white and w=colored. At the second gene yellow is dominant to green, and the symbols used are G=yellow, g=green. If the dihybrid is selfed, three phenotypes are produced in a 12:3:1 ratio.

The following table explains how this ratio is obtained.

Shapes of Squash Fruit :

Genotype	Fruit Color	Gene Actions
9 W_G_	White	Dominant white allele negates effect of G allele
3 W_gg	White	Dominant white allele negates effect of G allele
3 wwG_	Yellow	Recessive color allele allows yellow allele expression
1 wwgg	Green	Recessive color allele allows green allele expression

Because the presence of the dominant W allele masks the effects of either the G or g allele, this type of interaction is called dominant epistasis.

Example 4: 13:3 ratio
Phenotype: Malvidin production in *Primula*

Certain genes have the ability to suppress the expression of a gene at a second locus. The production of the chemical malvidin in the plant Primula is an example. Both the synthesis of the chemical (controlled by the K gene) and the suppression of synthesis at the K gene (controlled by the D gene) are dominant traits. The F1 plant with the genotype KkDd will not produce malvidin because of the presence of the dominant D allele. What will be the distribution of the F2 phenotypes after the F1 was crossed?

Genotype	Phenotype and genetic explanation
9 K_D_	no malvidin because dominant D allele is present
3 K_dd	malvidin productions because dominant K allele present
3 kkD_	no malvidin because recessive k and dominant D alleles present
1 kkdd	no malvidin because recessive k allele present

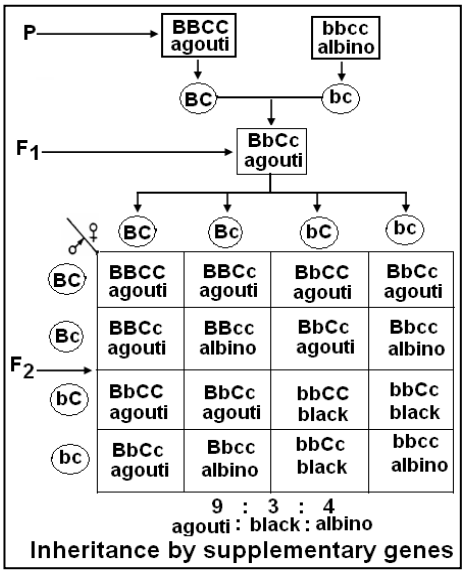
The ratio from the above table is 13 no malvidin production to 3 malvidin production. Because the action of the dominant D allele masks the genes at the K locus, this interaction is termed dominant suppression epistasis.

Suppressor - a genetic factor that prevents the expression of alleles at a second locus; this is an example of epistatic interaction. Remember that epistasis is the interaction between different genes. If one allele or allelic pair masks the expression of an allele at the second gene, that allele or allelic pair is epistatic to the second gene. Therefore, the following table summarizes the four epistatic interactions discussed above.

Example	Allelic Interactions	Type of Epistasis
Wheat kernel color	A epistatic to B,b B epistatic to A,a	Duplicate genes
Sweet pea flower color	cc epistatic to P,p pp epistatic to C,c	Complementary gene action
Squash Fruit Color	W epistatic to G, g	Dominant epistasis
Primula malvidin production	D epistatic to K, k	Dominant suppression

Supplementary Gene Interaction:

- ✓ Involves 2 pairs of non-allelic genes
- ✓ Affect the same character
- ✓ One of the dominant gene has visible effect itself
- ✓ Second dominant gene expresses itself when supplemented by the other dominant gene of pair
- ✓ Coat color (black, albino and agouti) of mice follows supplementary gene interaction.



Inheritance by supplementary genes

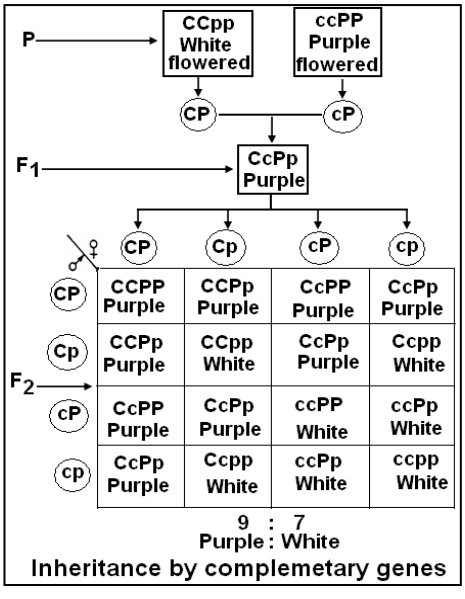
In mice, black coat color is monogenically dominant over albino and agouti. The offspring resulting from the cross between black and albino has agouti color.

F_2 generation shows segregation in the ratio 9 agouti: 3 black: 4 albino. This behavior is based on ratio of dihybrid cross, so the trait must be governed by two pairs of genes.

Suppose, gene C is essential for the development of black coat color, so present in black mice and absent in albino mice. Albino mice contains only gene B, so produces albino phenotype. But, when gene B is present along with gene C, produces agouti phenotype. Both the genes in recessive form produce albino phenotype.

Complementary Gene Interaction:

- ✓ Involves two pairs of non-allelic genes
- ✓ When dominant forms of both the genes involved in complementary gene interaction are alone have same phenotypic expression
- ✓ But, if they are present in combination, yield different phenotypic effect.
- ✓ Flower color follows this type of gene interaction.



Inheritance by complemetary genes

We have considered garden pea for the explanation of this type of gene interaction. Two different varieties of garden pea produce same color flowers – white. But on crossing they yield purple color flowers. Again in F_2, 9 purple : 7 white segregation was observed. How this happened? The answer is complementary gene interaction.

Suppose, Gene W in variety I and gene C in variety II produces white flowers. But, the progeny resulting from cross between these two has purple flowers. This means both the dominant genes, W and C together produce purple color flowers. In the variety I, dominant C gene is absent but it posses recessive c gene. Similarly, variety II contains recessive w gene. Both recessive genes produce white flowers, when present together. As in this type of interaction, the two recessive genes complement each other; it is called as complementary gene interaction.

Cross between			**F_1**	**F_2**
CCpp	X	ccPP	CcPp	9 purple : 7 white
white	X	white	purple	

Multiple Alleles: An **allele** is one of two or more forms of the DNA sequence of a particular gene. Each gene can have different alleles. Sometimes, different DNA sequences (alleles) can result in different traits, such as color. Sometimes, different DNA sequences (alleles) will have the same result in the expression of a gene. Most multicellular organisms have two sets of chromosomes, that is, they are diploid. These chromosomes are referred to as homologous chromosomes. Diploid organisms have one copy of each gene (and one allele) on each chromosome. If both alleles are the same, they are homozygotes. If the alleles are different, they are heterozygotes.

Characters of Multiple Alleles: are given below:

1. Multiple alleles of a series occupy the same locus in the chromosome.
2. Because, all the alleles of multiple series occupy same locus of chromosomes, therefore, no crossing over within the alleles of a same multiple allele series.
3. Multiple alleles always influence the same character.
4. The wild type allele is nearly dominant, while the other mutant alleles in the series may show dominance or there may be intermediate phenotypic effect.
5. When any of two of the mutant multiple alleles are crossed, phenotype is mutant type and not the wild type.

Dominant and recessive alleles: In many cases, genotypic interactions between the two alleles at a locus can be described as dominant or recessive, according to which of the two homozygous genotype the phenotype of the heterozygote most resembles. Where the heterozygote is indistinguishable from one of the homozygotes, the allele involved is said to be dominant to the other, which is said to be recessive to the former. The degree and pattern of dominance varies among loci.

Allele and genotype frequencies: The frequency of alleles in a population can be used to predict the frequencies of the corresponding genotypes. For a simple model, with two alleles: Where p is the frequency of one allele and q is the frequency of the alternative allele, which necessarily sum to unity. Then, p^2 is the fraction of the population homozygous for the first allele, $2pq$ is the fraction of heterozygotes, and q^2 is the fraction homozygous for the alternative allele. If the first allele is dominant to the second, then the fraction of the population that will show the dominant phenotype is $p^2 + 2pq$, and the fraction with the recessive phenotype is q^2.

Coat Colour in Rabbits: is controlled by multiple alleles. There are as follows.

[1] Full colour: The coat of the ordinary (wild type) rabbit is referred to as "agouti" or full colour, in which individuals have banded hairs, the portion nearest the skin being gray, succeeded by a yellow band, and finally a black or brown tip. The allele for full colour may be represented by capital letter c^+.

[2] Chinchilla: In some individuals, the coat lacking the yellow pigment and due to the optical effect of black and gray hairs, have the appearance of silvery-gray. The allele for chinchilla is represented as c^{ch}

[3] Himalyan (Russian): The Himalyan type coat is white except for black extremities (nose, ears, feet and tail). The condition in which black pigmentation is confined to the ears, muzzle, feet and tail, is called acromelanism (Serra, 1965). In Himalyan rabbits eyes remain pigmented. The allele for Himalyan coat is represented by c^h.

[4] Albino: The albino coat totally lacks in pigmentation and the eyes of a albino also remain pink due to lack of pigment in iris of eye. Allele for albino is represented by c.

Crosses of homozygous agouti (c+c+) and albino (cc) individuals produce a uniform agouti F1; interbreeding of the F1 produces an F2ratio of 3 agouti: 1 albino. Two third of F2 agouti are found to be heterozygous by testcrosses. Thus, it is a case of monohybrid inheritance, with agouti completely dominant to albino.

Likewise, crosses between chinchilla and agouti produce all agouti individuals in the F1 and a 3 agouti: 1 chinchilla ratio in the F2. Such complete dominance of agouti also occurs on Himalayan. Further crosses, reveal that cch allele for chinchilla, though is recessive to c+ allele for agouti coat or skin is incompletely dominant over Himalayan (ch) and albino (c) alleles. Likewise, ch allele for Himalayan coat is recessive to c+ (agouti) and cch (chinchilla) but dominates over albino. The results of all these crosses exhibit that c+ (agouti), cch (chinchilla), ch (Himalayan) and c (albino) are allelic to each other and the alleles of this multiple allelic series have following dominance hierarchy : $c^{+}>c^{ch}>c^{h}>c$ The possible phenotypes and their associated genotypes of this multiple allelic series can be summarized below.

P_1:	Agouti	X	Albino
	$C^{+}C^{+}$	↓	CC
		Agouti	
F_1:		$C^{+}C$	
F_2:		$1c^{+}c^{+}:2c^{+}c:1cc$	
		3 Agouti : 1 Albion	

P_1:	Agouti	X	Chinchilla
	$C^{+}C^{+}$	↓	$C^{ch}C^{ch}$
		Agouti	
F_1:		$C^{+}C^{ch}$	
F_2:		$1c^{+}c^{+}:2c^{+}c^{ch}:1c^{ch}c^{ch}$	
		3 Agouti : 1 Chinchilla	

P_1:	Agouti	X	Himalayan
	$C^{+}C^{+}$	↓	$C^{h}C^{h}$
F_1:		Agouti	
		$C^{+}C^{h}$	
F_2:		$1\ C^{+}C^{+}:2C^{+}C^{h}:1\ C^{h}C^{h}$	
		3 Agouti : 1 Himalayan	

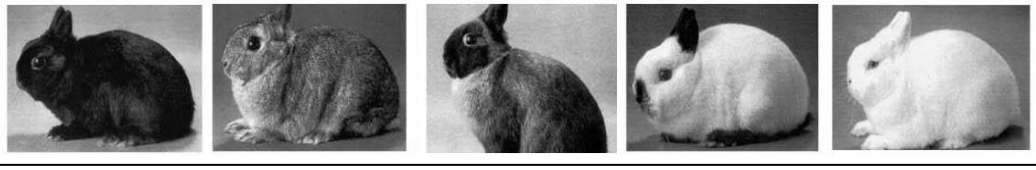
P_1:	Chinchilla	X	Himalayan
	$C^{ch}C^{ch}$	↓	$C^{h}C^{h}$
F_1:		Light gray	
		$C^{ch}C^{h}$	
F_2:		$1C^{ch}C^{ch}: 2C^{ch}C^{h}:1\ C^{h}C^{h}$	
		1 Chinchilla : 2 Light gray:1 Himalayan	

P_1:	Chinchilla	X	Albino
	$C^{ch}C^{ch}$	↓	C C
F_1:		Light gray	
		$C^{ch}C$	
F_2:		$1C\ ^{ch}C^{ch}:2\ C^{ch}C\ :1cc$	
		1 Chinchilla : 2 Light gray : 1 Albino	

P_1:	Himalayan	X	Albino
	$C^{h}C^{h}$	↓	C C
F_1:		Himalayan	
		$C^{h}C$	
F_2:		$1\ C^{h}C^{h}:\ 2C^{h}C\ :\ 1\ C\ C$	
		3 Himalayan : 1 Albino	

The phenotypes and genotypes of multiple allelic series for coat colour in rabbit.

Possible genotypes	CC, Cc^{ch}, Cc^{h}, Cc	$c^{ch}c^{ch}$	$c^{ch}c^{h}, c^{ch}c$	$c^{h}c^{h}, c^{h}c$	cc
Phenotype	Dark gray	Chinchilla	Light gray	Himalayan	Albino



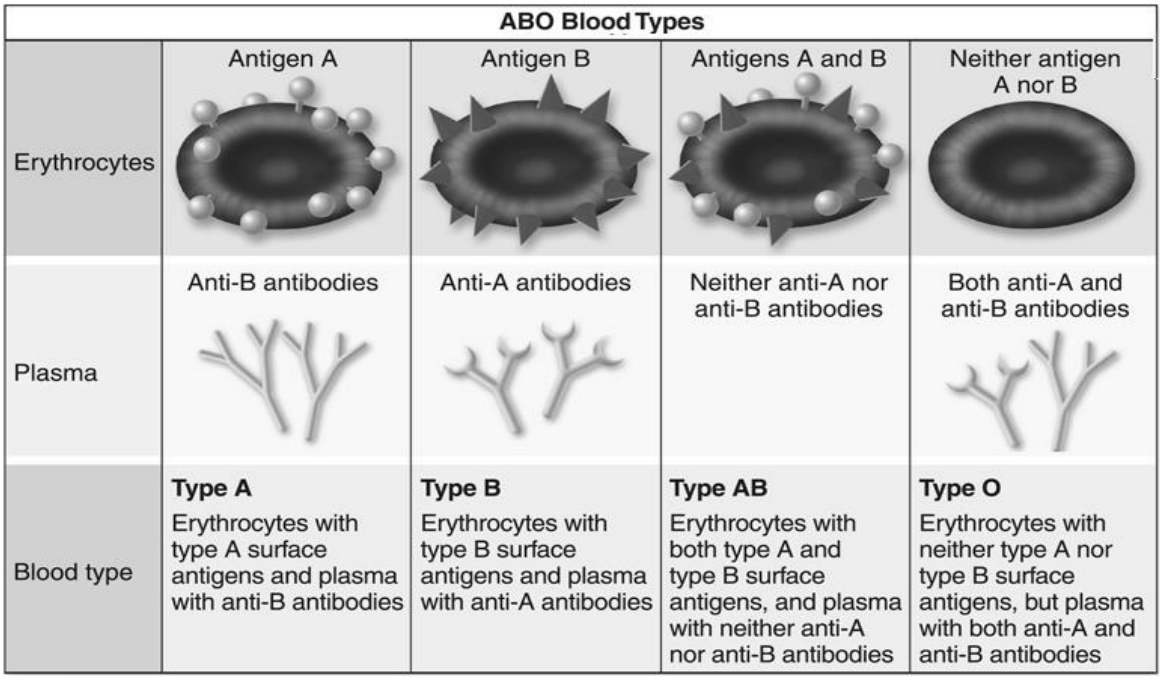
ABO Blood group in man: Multiple Alleles inheritance of ABO blood group types of man is determined by a series of three allelomorphic genes.

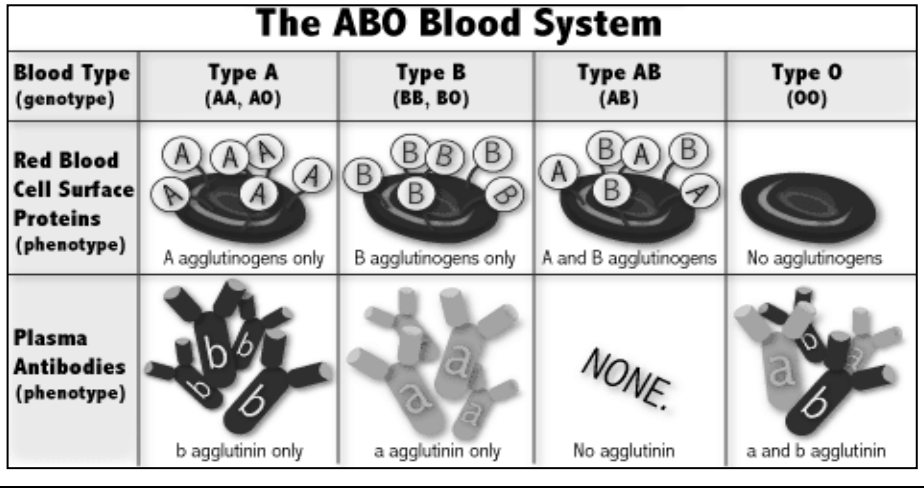
For example, at the gene locus for ABO blood type proteins in humans, classical genetics recognizes three alleles, IA, IB, and IO, that determines compatibility of blood transfusions.

Any individual has one of six possible genotypes (AA, AO, BB, BO, AB, and OO) that produce one of four possible phenotypes:

1) "A" - produced by AA homozygous and AO heterozygous genotypes
2) "B" - produced by BB homozygous and BO heterozygous genotypes
3) "AB" - heterozygotes
4) "O" - homozygotes.

It is now appreciated that each of the A, B, and O alleles is actually a class of multiple alleles with different DNA sequences that produce proteins with identical properties: more than 70 alleles are known at the ABO locus. An individual with "Type A" blood may be a AO heterozygote, an AA homozygote, or an A'A heterozygote with two different 'A' alleles.





Rh blood group system: The **Rh (Rhesus) blood group system** (including the **Rh factor**) is one of the currently 30 human blood group systems. It is clinically the most important blood group system after ABO. The Rh blood group system currently consists of 50 defined blood-group antigens, among which the 5 antigens D, C, c, E, and e are the most important ones. The commonly-used terms *Rh factor*, *Rh positive* and *Rh negative* refer to the *D antigen* only. Besides its role in blood transfusion, the Rh blood group system, in particular the D antigen, is a relevant cause of the hemolytic disease of the newborn or erythroblastosis fetalis for which prevention is key.

Rh factor: Individuals either have, or do not have, the "*Rhesus factor*" on the surface of their red blood cells. This term strictly refers only to the most immunogenic D antigen of the Rh blood group system, or the Rh- blood cell system. The status is usually indicated by *Rh positive* (Rh+, does have the D antigen) or *Rh negative* (Rh-, does not have the D antigen) suffix to the ABO blood type.

However, other antigens of this blood group system are also clinically relevant. These antigens are listed separately. In contrast to the ABO blood group, immunization against Rh can generally only occur through blood transfusion or placental exposure during pregnancy.

Rh nomenclature: The Rh blood group system has two sets of nomenclatures: one developed by Fisher and Race, the other by Wiener. Both systems reflected alternative theories of inheritance. The Fisher-Race system, which is more commonly in use today, uses the CDE nomenclature.

This system was based on the theory that a separate gene controls the product of each corresponding antigen (e.g., a "D gene" produces D antigen, and so on). However, the d gene was hypothetical, not actual.

The Wiener system used the Rh-Hr nomenclature. This system was based on the theory that there was one gene at a single locus on each chromosome, each contributing to production of multiple antigens. In this theory, a gene R_1 is supposed to give rise to the “blood factors” Rh_0, rh’, and hr” (corresponding to modern nomenclature of the D, C and e antigens) and the gene r to produce hr’ and hr” (corresponding to modern nomenclature of the c and e antigens).

Notations of the two theories are used interchangeably in blood banking (e.g., Rho(D) meaning RhD positive). Wiener's notation is more complex and cumbersome for routine use. Because it is simpler to explain, the Fisher-Race theory has become widely used.

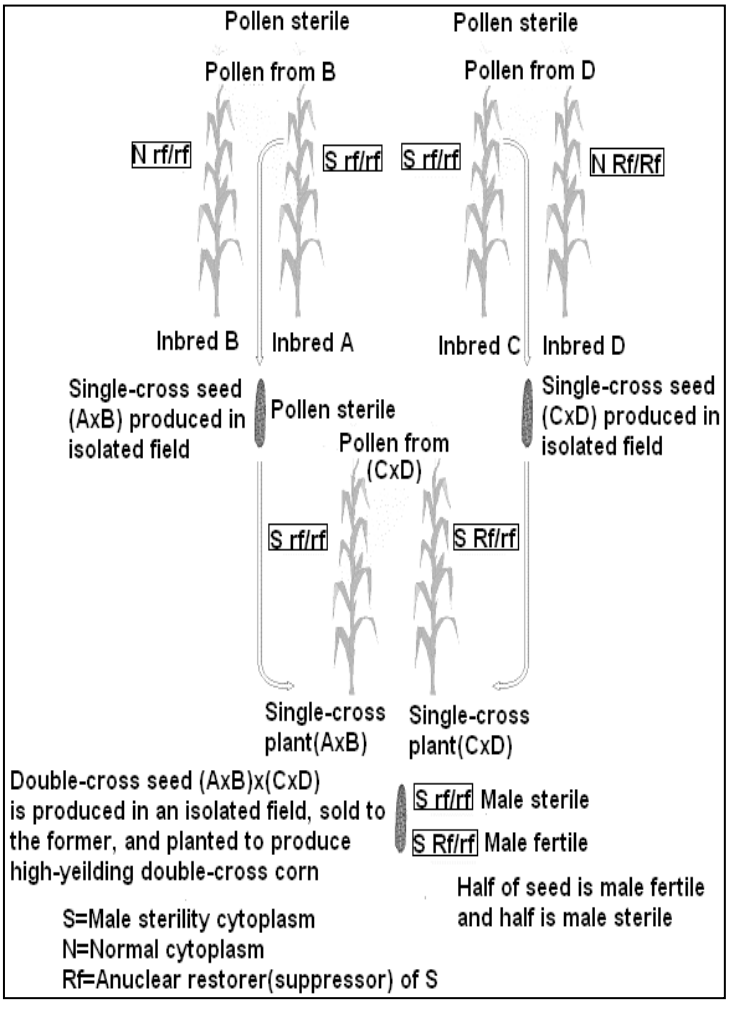
Inheritance: The D antigen is inherited as one gene (*RhD*) (on the short arm of the first chromosome, p36.13-p34.3) with various alleles. As very much simplified, one can think of alleles that are positive or negative for the D antigen. The gene codes for the RhD protein on the red cell membrane. D- individuals who lack a functional *RHD* gene do not produce the D antigen, and may be immunized by D+ blood.

The epitopes for the next 4 most common Rh antigens, C, c, E and e are expressed on the highly similar RhCE protein that is genetically encoded in the *RhCE* gene. It has been shown that the *RhD* gene arose by duplication of the *RhCE* gene during primate evolution. Mice have just one RH gene.

Male sterility: Male sterility can be aroused spontaneously via mutations in nuclear and/or cytoplasmic genes. Male sterility can be either cytoplasmic or cytoplasmic-genetic. Cytoplasmic male sterility (CMS) is caused by the extranuclear genome (mitochondria / chloroplast) and shows maternal inheritance. Manifestation of male sterility in CMS may be either entirely controlled by cytoplasmic factors or by the interaction between cytoplasmic and nuclear factors.

Cytoplasmic male sterility: Cytoplasmic male sterility, as the name indicates, is under extra nuclear genetic control. They show non-Mendelian inheritance and are under the regulation of cytoplasmic factors. In this type, male sterility is inherited maternally. In general there are two types of cytoplasm: N (normal) and the aberrant S (sterile) cytoplasms. These types exhibit reciprocal differences.

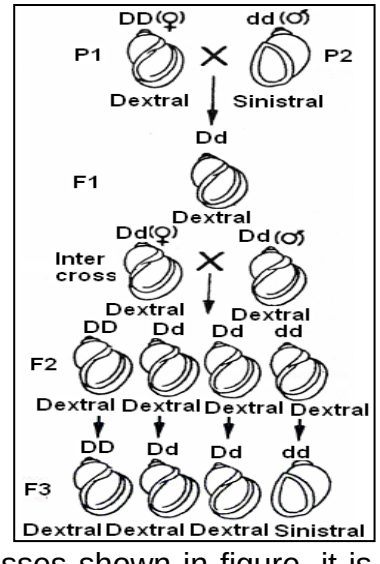
Cytoplasmic-genetic male sterility: While CMS is controlled by an extranuclear genome often times nuclear genes can have the capability to restore fertility. When nuclear restorations of fertility genes ("Rf") are available for CMS system in any crop, it is cytoplasmic-genetic male sterility; the sterility is manifested by the influence of both nuclear (Mendelian inheritance) and cytoplasmic (maternally inherited) genes. There are also restorers of fertility (Rf) genes, which are distinct from genetic male sterility genes. The Rf genes do not have any expression of their own unless the sterile cytoplasm is present. Rf genes are required to restore fertility in S cytoplasm which causes sterility. Thus N cytoplasm is always fertile and S cytoplasm with genotype Rf- produces fertiles; while S cytoplasm with rfrf produces only male steriles. Another feature of these systems is that Rf mutations are frequent, so N cytoplasm with Rfrf is best for stable fertility.



Cytoplasmic-genetic male sterility systems are widely exploited in crop plants for hybrid breeding due to the convenience to control the sterility expression by manipulating the gene–cytoplasm combinations in any selected genotype. Incorporation of these systems for male sterility evades the need for emasculation in cross-pollinated species, thus encouraging cross breeding producing only hybrid seeds under natural conditions.

Cytoplasmic male sterility in hybrid breeding: Cytoplasmic male sterility is used in hybrid seed production. In this case, the sterility is transmitted only through the female and all progeny will be sterile. This is not a problem for crops such as onions or carrots where the commodity harvested from the F1 generation is produced during vegetative growth. These CMS lines must be maintained by repeated crossing to a sister line (maintainer line) that is genetically identical except that it possesses normal cytoplasm and is therefore male fertile. In cytoplasmic-genetic male sterility restoration of fertility is done using restorer lines carrying nuclear restorer genes in crops. The male sterile line is maintained by crossing with a maintainer line which has the same genome as that of the MS line carries normal fertile cytoplasm.

Maternal Effects: Since chromosomes divide in a very precise manner both during mitosis as well as meiosis, it is easy to draw a parallelism between chromosomes and genes. Cytoplasm, however does not divide in such a manner during cell division. Female gamete usually contributes more cytoplasm to the zygote. Consequently, for characters having cytoplasmic control, differences in reciprocal crosses would be observed. Inheritance in these cases would be mainly of maternal type as shown in figure. If two strains A and B respectively having genotype AA and BB and cytoplasms a and b are crossed reciprocally, we will get two hybrids AB(a) and AB(b) (Cytoplasm is indicated in parentheses). In case of maternal effect, AB(a) and AB(b), despite having same nuclear genotype will differ. AB(a) will resemble strain A or AA(a) and AB(b) will resemble strain B or BB(b). since such effects are solely produced by cytoplasm of the egg, they are described as maternal effects. However, maternal effects are often produced due to effect of genes through cytoplasm. In other words, properties of cytoplasm depend on nuclear genes. Such cases can be distinguished from those, where extra-chromosomal or cytoplasmic heredity units are present and function either independently or in collaboration with nuclear genetic system. This is called extra-chromosomal or cytoplasmic or organellar inheritance, and is distinguished from maternal effects discussed above.



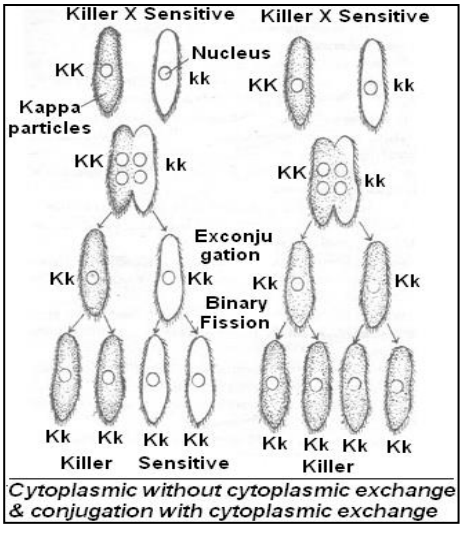
Coiling shell in snail (Limnea peregra): In the snails, the shell is spirally coiled. In most cases the direction of coiling of the shell is clockwise, if viewed from apex of the shell. This type of coiling is called dextral. However, in some snails the coiling of shell may be counter clockwise or sinistral. Both types of coilings are produced by two different types of genetically controlled cleavages, one being dextral cleavage, another being sinistral cleavage. The dextral coiling depending upon dominant allele D and sinistral coiling depending upon recessive allele d, so the dextral is DD and sinistral is dd.

The phenotype in progeny obtained from reciprocal crosses (♀DD X ♂dd : ♀dd x ♂DD) is determined by the genotype and not by the phenotype of female parent. In reciprocal crosses shown in figure, it is evident that the genotype Dd(F1) can be dextral as well as sinistral depending upon the genotype of female parent. Similarly, dd can be dextral if genotype of female parent carries dominant allele (Dd). It should be carefully noted that phenotype of female parent does not have any effect on phenotype of progeny. It is genotype of female parent which is really decisive. This is an example of delayed effect of genotype.

CO_2 sensitivity in Drosophila: Most Dorsophilia flies, can be subjected to contact with pure CO_2 for long hours, without injury. However a true breeding strain of Dorsophilia which was sensitive to CO_2. The sensitive flies, when exposed briefly to CO_2 for a short period, become unconscious in a characteristics way, with their legs becoming paralysed. When reciprocal crosses were made between CO_2 sensitive and normal strains, it could be shown that the trait was inherited only from female parent. In other words, while sensitive mothers always give sensitive progeny, sensitive fathers only rarely five sensitive progeny if the mother is normal.

In rare transmission through male also, sensitivity is quickly lost after first generation. It has also been shown that if an extract obtained by crushing sensitive flies, is injected into the body of wild (Normal) flies, sensitivity can be induced. It was also shown that this sensitivity can be attributed to a virus like particles called sigma found in cytoplasm of the cells of sensitive fly. Sigma factor is transmitted through egg cytoplasm and its reproduction depends on initial supply and on suitable temperature of 20^0C, because it is a heat liable at high temperature. It does not need a specific gene and may be found associated with different genotypes. A sensitive fly retains its sensitive trait, even when all its chromosomes are replaced by those of normal fly, suggesting that sigma factor or particle is located in cytoplasm and has properties of non-chromosomal genes or plasma genes. Some physical characteristics of sigma are also known. It seems to be a particle of 0.07 µ in diameter, and contains DNA responsible for its hereditary nature.

Kappa Particles in Paramecium: There are two strains of Paramecium. They are killer and sensitive. Killer strain produces a toxic substance called paramecin that kills the other type. The production of paramecin in killer type is controlled by certain cytoplasmic particles known as kappa particles. The sensitive strains lack these particles. The kappa particles pass from one generation to the next generation in the process of cell division. The kappa particles are also multiplied with cell division. They are transmitted through the cytoplasm. But their multiplication is controlled by a dominant nuclear gene K. Kappa is dependent for its continued existence on K. The gene K can only maintain the kappa particles and cannot initiate its production.

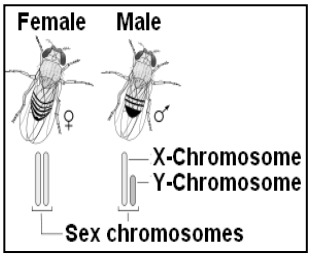


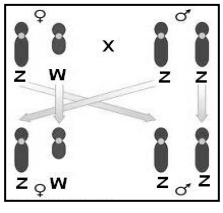
Cytoplasmic without cytoplasmic exchange & conjugation with cytoplasmic exchange

When killers KK conjugate with non-killers kk, the exconjugants are Kk. But the development of a particular type depends upon the duration of cytoplasmic exchange. In normal case of conjugation, the nuclear material alone is exchanged and there is no exchange of cytoplasmic material. In such cases, each exconjugant gives rise to the organisms of its own type. i.e., killer exconjugant produces killers and non-killer produces non-killers. Sometimes the conjugation period is prolonged and the cytoplasmic bridge between the two conjugants is larger. In such cases, in addition to the nuclear material, the cytoplasmic materials are also exchanged. During this cytoplasmic exchange, the kappa particles present in the cytoplasm of the killer type enter the non-killer type and convert it into a killer type. So all the offspring produced by the exconjugants are killer type. This shows that a Paramecium becomes a killer when it receives kappa particles & becomes a sensitive when it doesn't receive kappa particles.

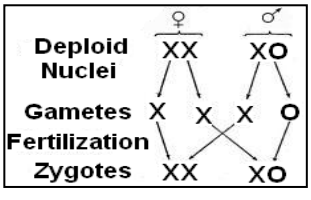
Sex Determination: In dioecious species (separate sexes) there are several means to determine sex. The chromosomes involved in sex determination are called sex chromosomes. All other chromosomes are called autosomal chromosomes or autosomes. Although sex chromosomes provide the most common means of sex determination, it is not the only mechanism. Chromosomal sex determining mechanisms as this is most common and is the mechanism seen in mammals.The autosomes occur in homologous pairs with each chromosome possessing one copy (allele) of each gene. Segregation and reassortment lead to the pattern of inheritance, which is called Mendelian inheritance. The sex chromosomes may be genetically distinct thus homologous pairs may not exist and this leads to inheritance patterns that are different from autosomal inheritance. There are four basic types of chromosomal mechanisms

1. **XX-XY** in which females are homomorphic XX and males are heteromorphic XY. This is found in mammals including humans and some insects including *Drosophila*. In humans, females have 23 homomorphic pairs and males have 22 homomorphic pairs plus a heteromorphic pair. During meiosis, females produce only one kind of gamete all having one X chromosome. Males produce two kinds of gametes, one with an X and the other with a Y chromosome. Females are homogametic and males are heterogametic.





2. **ZZ-ZW** system in which females are heteromorphic ZW and males are homomorphic ZZ. This occurs in birds, some fishes, and moths. It is essentially the opposite of XY in mammals.



3. **XX-XO** system in which females have 2 X chromosomes. Males have only 1 X and no additional sex chromosomes. Females have an even number of chromosomes and males have an odd number of chromosomes. This occurs in many species of insects. This was the first sex determining mechanism discovered, and the sex determining chromosome was named the X in 1905. Gametes of males have either an X chromosome or no sex chromosome.

4. **Compound** chromosome system: These can be very complex with multiple numbers of X and Y chromosomes. e.g. in *Ascaris incurva*, a nematode, there are 26 autosomes, 8 X chromosomes, and 1 Y chromosome Males have 26A + 8X + Y for 35 chromosomes Females have 26A + 16X for 42 chromosomes This type of system is also common in spiders. Sex determination in the XY system is the most studied because it is found in humans and *Drosophila*. It varies from species to species. In *Drosophila*, the greater the number of X chromosomes relative to the autosomes, the more likely the individual will be female (table).

Phenotype	**Chromosomal complement**	**# of X# of autosomal sets**
Normal female	XX + 2N autosomes	1.00
Normal male	XY + 2N autosomes	0.50
Metafemale	XXX + 2N autosomes	1.50
Metamale	X + 3N autosomes	0.33
Intersex	XX + 3N autosomes	0.67

Chromosomal Theory of Sex Determination: According to this theory, the chromosome is the main factor to determine the sex. The chromosomal theory of sex determination was proposed by *Mc Clung.*

There are two of chromosomes in an organism. They are the **autosomes** and **allosomes.** The autosomes contain genes which determine the somatic characters of the organisms. The allosomes determine, the sex of an organism. Hence the allosomes are also called **sex chromosomes.** There are two types of sex chromosomes. They are **X** chromosome and **Y** chromosomes. These two chromosomes differ not only in appearance but also in genetic composition.

The **X** chromosome is larger in size and is straight. It contains a large amount of euchromatin and a small amount of heterochromatin. The euchromatin is rich DNA. So the **X** chromosome carries large amount of DNA (gene).

The **Y** chromosome is smaller in size. The Y chromosome of Drosophila has a bent at one end. But the Y chromosome of man is straight. It contains a small amount of euchromatin and a large amount of heterochromatin. Hence the amount of DNA in Y chromosome is less. As a result, the Y chromosome is genetically **inert** of **inactive.**

In a normal animal, there are two sex chromosomes. The two sex chromosomes are **XX** or **XY.** In man, insects, etc. the female has two **X** chromosomes and the male has one **X** chromosome and one **Y** chromosome. The chromosomal theory of sex determination is subdivided into the following types:

1. *Theory of heterogamesis*
2. *Genic balance theory*
3. *Quantitative theory*
4. *Haploid-diploid mechanism*
5. *Gynandromorphs*
6. *Cytological basis and*
7. *Monofactorial sex determination.*

Genic Balance Theory: According to genic balance theory the sex is determined by the ratio between X chromosomes and autosomes. This theory was formulated by Bridges. According to this theory, sex is determined by the relative number of X chromosomes and autosomes. It is actually the ratio between the X chromosomes and autosomes determines the sex. The X chromosomes carry female stimulating genes and the autosomes (A) seem to carry the male stimulating genes. There is no sex influencing genes in Y chromosomes. Haploid sets of autosomes are represented as n (A) and diploid sets of autosomes are represented as 2n(A). The sex of an animal is determined by the ratio between the number of X chromosomes and the number of haploid sets of autosomes. The ratio is the quantitative balance between X chromosomes and autosomes.

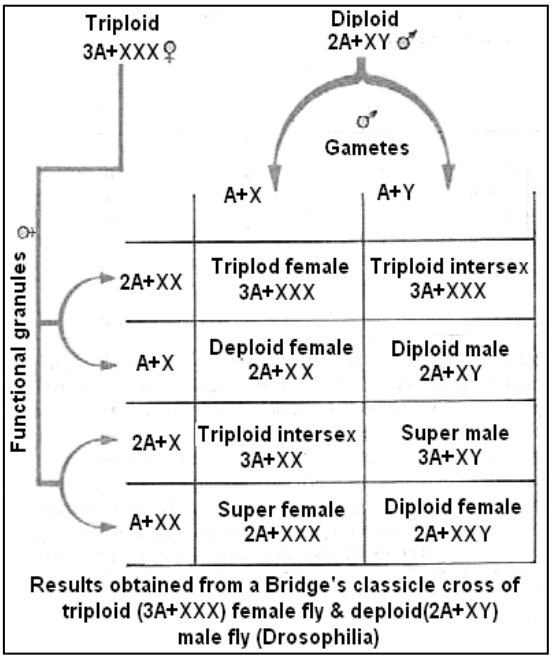
$$\text{Sex determining ratio (Sex Index)} = \frac{\text{Number of X chromosomes}}{\text{Number of haploid sets of autosomes}}$$

If the sex index is 1 the individual develops into female. If the sex index is 0.5, it develops into male. If the ratio is intermediate (0.67) between 1 and 0.5 the resulting

individual is an intersex. If the ratio is above 1 (1.5) the sex is super female and if the ratio is below 0.5 (0.3), the sex is supermale.

In male Drosophila, there are 2 sets of autosomes 2n(A) and one X chromosome. Hence the ratio is X/2n(A)+1/2 = 0.5. In female, there are two sets of autosomes 2n(A) and two X chromosomes. Hence the ratio is 2/2 = 1.

Bridges also explained the formation of supersexes and intersexes in Drosophila. He found some Drosophila females with triploid sets of chromosomes 3n(A) XXX. These triploid females are much like the normal diploid ones in appearance and are fertile. Bridges crossed this triploid female with normal diploid male. The diploid normal male produces two types of sperms. The triploid female produces four types of eggs. When the four types of eggs are fertilized by two types of sperms, eight sexually distinct kinds of offspring are produced as in the checker board.



Results obtained from a Bridge's classicle cross of triploid (3A+XXX) female fly & deploid(2A+XY) male fly (Drosophilia)

Triploid intersexes in drosophila: Bridges crossed the experimentally produced triploid (3n individual having three whole sets of chromosomes) female Dorsophilia (3A:3X) to diploid males (2A:XY). The results obtained from such a cross are shown in figure. From this cross he obtained normal diploid males, triploid females, intersexes, super males and super females. The occurrence of triploid intersexes from such a cross, clearly established that the autosomes also carry genes for sex determination. These intersexes were sterile individuals and had a phenotype in between male and female sexes, the occurrence of such intersexes, super males and super females were explain by him by genic balance mechanism. Different combination of X chromosomes and autosomes and corresponding sex expression in Dorsophilia can be summarized in table.

As shown in the table, when the X/A ratio is 1.0, the individual will be female and if it is 0.50, it could be male, when this balance is disturbed, the sex of individual deviate from normal male or normal female. For example, when the X/A ratio falls between 1.0, and 0.50, it would be intersex; when it is below 0.50 it would be super male and when above 1.0, it would be meta female of super female.

No.	Nature of the sex	Number of chromosomes	Number of Autosomes (A Sets)	Sex Index = No. X's/No. A Sets
1.	Super female	3X	2n(A)	3/2 = 1.5
2.	Tetraploid female	4X	4n(A)	4/4 =1.0
3.	Triploid female	3X	3n(A)	3/3 = 1.0
4.	Diploid female	2X	2n(A)	2/2 = 1.0
5.	Haploid female	1X	1n(A)	1/1 =1.0

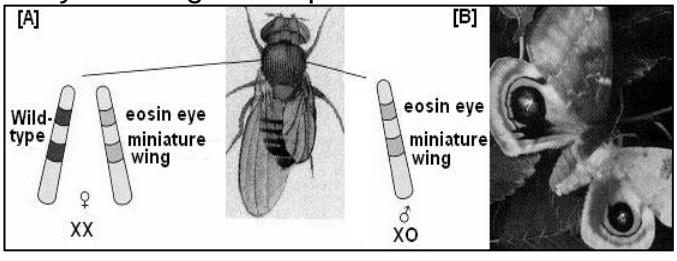
6.	Intersex	2X	3n(A)	2/3 = 0.67
7.	Normal male	1X	2n(A)	1/2 = 0.50
8.	Super male	1X	3n(A)	1/3 = 0.33

Gynandromorphs in Drosophila melanogaster: In Drosophila, and in insects in general, one can observe gynandromorphs animals in which certain regions of the body are male and other regions are female. This can happen when an X chromosome is lost from one embryonic nucleus. The cells descended from that cell, instead of being XX (female), are XO (male). Because there are no sex hormones in insects to modulate such events, each cell makes its own sexual "decision." The XO cells display male characteristics, whereas the XX cells display female traits. This situation provides a beautiful example of the association between insect X chromosomes and sex.

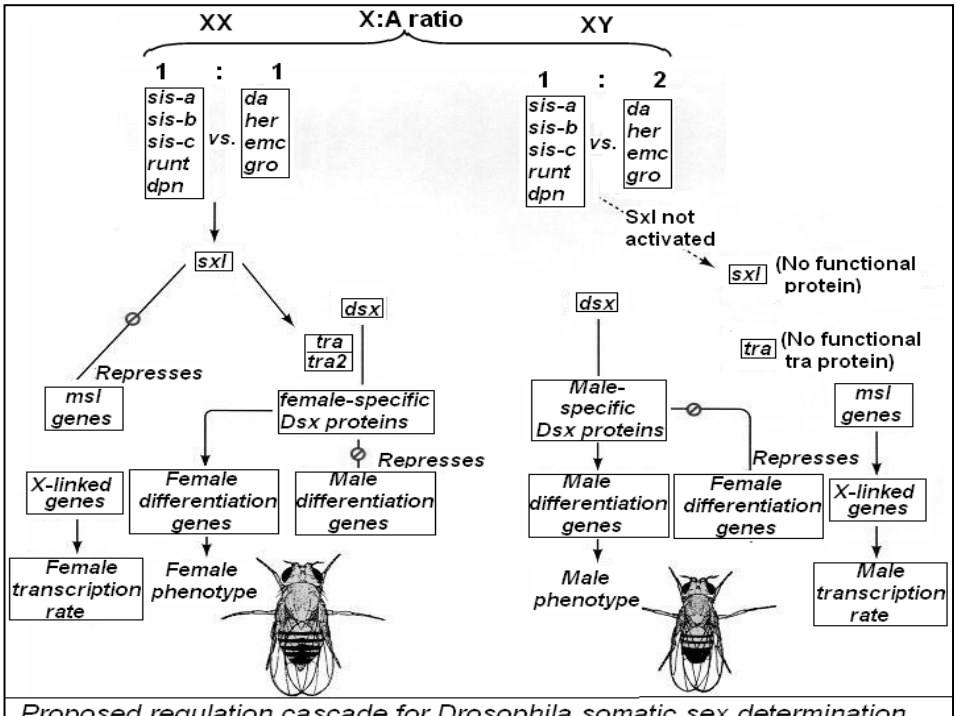
Gynandromorphs:

(A) Gynandromorph of *D. melanogaster* in which the left side is female (XX) and the right side is male (XO). The male side has lost an X chromosome bearing the wild-type alleles of eye color and wing shape, thereby allowing the expression of the recessive alleles eosin eye and miniature wing on the remaining X chromosome.

(B) Photograph of a gynandromorphic of moth, divided bilaterally into a rose-brown female half and a smaller, yellow male half.



X/A ratio in drosophila: Any theory of *Drosophila* sex determination must explain how the X-to-autosome (X:A) ratio is read and how this information is transmitted to the genes controlling the male or female phenotypes. Several genes with roles in sex determination have been found. Loss-of-function mutations in most of these genes *Sex-lethal* (*Sxl*), *transformer* (*tra*), and *transformer-2* (*tra2*)—transform XX individuals into males. Such mutations have no effect on sex determination in XY males. Homozygosity of the *intersex* (*ix*) gene causes XX flies to develop an intersex phenotype having portions of male and female tissue in the same organ. The *doublesex* (*dsx*) gene is important for the sexual differentiation of both sexes. If *dsx* is absent, both XX and XY flies turn into intersexes.



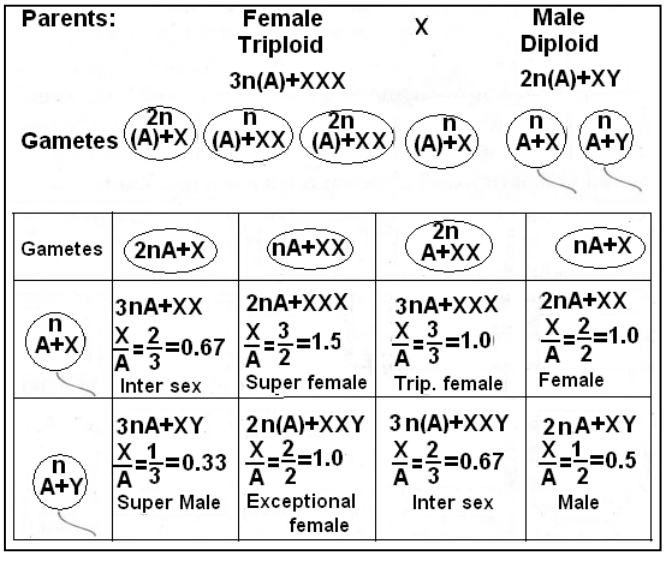
Proposed regulation cascade for Drosophila somatic sex determination. Arrows represent activation, while a block at the end of a line indicates suppression. The msl loci, under the control of the Sxl gene, regulate the dosage compensatory transcription.

The positioning of these genes in a developmental pathway is based on

1) The interpretation of genetic crosses resulting in flies bearing two or more of these mutations and
2) The determination of what happens when there is a complete absence of the products of one of these genes. Such studies have generated the model of the regulatory cascade seen in Figure

Table: Sexes and sex index of Drosophila.

No.	Nature of the sex	Number of chromosomes	Number of sets of Autosomes	Sex Index RatioX/A
1.	Super female	3X	2n(A)	3/2 = 1.5
2.	Triploid female	3X	3n(A)	3/3 = 1
3.	Diploid female	2X	2n(A)	2/2 = 1
4.	Intersex	2X	3n(A)	2/3 = 0.67
5.	Intersex	2XY	3n(A)	2/3 = 0.67
6.	Normal male	X	2n(A)	1/2 = 0.5
7.	Super male	X	3n(A)	1/3 = 0.33
8.	Exceptional female	2XY	2n(A)	2/2 = 1



Mutations Introduction: A mutation is any change in the sequence of the DNA encoding a gene. Most of these mutations are recognized because the phenotype of the organism has changed. Mutations have lead to a better understanding about the nature of genes. Originally genes were thought to be beads-on-string, where each bead was a single entity responsible for a phenotype. This theory leads to the concept that only single mutation was possible for a specific gene. Detailed genetic experiments proved that the gene actually consists of many individual units, and specific changes in these units can lead to several mutant phenotypes. Mutation is defined as a chemical change in the DNA structure of a gene. A difference of a single base in the DNA molecule or a single error in the reading of the code can cause a change in the amino acid sequence which leads to mutation. The chemical substances that induce mutations are known as mutagens. Mutation leads to the formation of defective genes, which causes abnormalities or diseases. Mutations are of following major types:

Types of Mutations

Classification of Detection of Mutations: A classification based on the method of detection of mutations includes the following main types:

1) **Morphological Mutations:** involves alterations in external form including color, shape, size, etc. examples include albino ascospores in *Neurospora,* kernel colour in corn, curly wings in *Dorsophilia* and dwarfism in pea.
2) **Lethal Mutations:** involve genotypic changes leading to death of an individual. These are perhaps easiest to score for study of mutation frequiences: some albino mutations resulting from chlorophyll deficiency are also lethal.
3) **Biochemical Mutations:** are identified by a deficiency, so that the defect can be overcome by supplying the nutrient or any other chemical compound, for which the mutant is deficient. Such mutations have been studied mainly in prokaryotes like bacteria and fungi, but sometimes also in eukaryotes like *Dorsophilia* and humans.
4) **Resistant Mutations:** are identified by their ability to grow in the presence of an antibiotic (e.g. streptomycin, ampicillin, cyclohexamide) or a pathogen, to which wild type is susceptible.
5) **Conditional Mutations:** are those which allow the mutant phenotype to express only under certain condition (e.g. temperature) called restrictive condition. Under other or normal condition described as permissive condition, the mutant expressed normal phenotype. These muatants, if lethal or semi-lethal can be multiplied under permissive conditions for specific study. They have been extensively used for study of cell cycle or for a study of DNA replication.

Classification of Mutations According to types of cells: According to their occurrence in somatic & germinal cell following types of mutations have been classified:

A. **Somatic Mutations:** The mutations occurring in non-reproductive body cells are known as somatic mutations. The genetical and evolutionary consequences of somatic mutations are insignificant, since only single cells and their daughter cells are involved. If, however, a somatic mutation occurs early during embryonic life, the mutant cells may constitute a large proportion of body cells and animal body may be

a mosaic for different types of cells somatic mutations have been often related with malignant growth. Examples of somatic mutation have been reported in *Oenothera lamarckiana a*nd several other cases including man.

B. Gametic Mutations: The mutations occurring in gamete cells (e.g. sperms and ova) are called gametic mutations. Such mutations are heritable and of immerce genetical significance. The gametic mutations only from the raw material for the natural selection.

Classification of Mutations According to types of cells: According to size following types of mutations have been categorized:

A. Point Mutations: When heritable alterations occur in a very small segment of DNA molecules, i.e. single nucleotide or nucleotide pair, then this type of mutations are called "point mutation". The point mutations may occur due to following types of subnucleotide change in the DNA and RNA.

1) Deletion Mutations: The point mutation which is caused due to loss or deletion of some portion of (single nucleotide pair) in a triplet codon of a gene is called deletion mutation. This have been reported in some bacteriophages.

2) Insertion Mutations: The point mutations which occur due to addition of one or more extra nucleotides to a gene or criston are called insertion mutations. The insertion mutations can be artificially induced by certain chemical substances called mutagens such as acridine dye and proflavin. A proflavin molecule, it is believed, insert between two successive bases of a DNA strand, thereby stretching the strand lengthwise. At replication, this situation would allow the insertion of an extra nucleotide in the complementary chain at the position occupied by the proflavin molecule. The mutation which arise from the insertion or deletion of individual nucleotides and cause the rest of the message downstream of the mutation to be read out of phase are called frameshift mutations.

3) Substitution Mutations: The point mutations in which a nucleotide of a triplet is replaced by another nucleotide, is called substitution mutation. The substitution mutation affect only a particular triplet codon.

B. Multiplication Mutations or Gross Mutations: When changes involving more than one nucleotide pair, or entire gene, then such mutation are called gross mutation. The gross mutation occur due to rearrangements of genes within the genome and may be of following types.

1) The rearrangement of genes may occur within gene. Two mutations within the same gene can produce different effects depending on gene wheather they occur in the cis or trans position.

2) The rearrangement of gene may occur in number of genes per chromosome. If the number of genes replicas is non-equivalent on the homologus chromosomes, they may cause different types of phenotypic effects over the organisms.

1) Due to movement of a gene locus new type of phenotype may be created, especially when the gene is relocated near heterochromatin. The movement of gene loci may take place due to following methods:

- **Translocation**: Movement of a gene may take place to a non-homologus chromosome and this is known as translocation.
- **Inversion**: The movement of a gene within the same chromosome is called inversion.

Classification of Mutations According to the Origin: Mutations are of two types: Spontaneous mutations and Induced mutations. According to size following types of mutations have been categorized:

Spontaneous Mutations: Mutations that are not due to external factors are called spontaneous mutations. Spontaneous mutations can occur at any point of the cell cycle. The mutation rate varies from ~10^{-4} to 10^{-6} mutations per gene per generation. The spontaneous mutation occurs suddenly in the nature and their origin is unknown, it is difficult to identify and score them. They are also called "*background mutation"* and have been reported in many organisms such as, *Oenothera, maize, bread molds, microorganisms (bacteria & viruses), Drosophilia, mice, man,* etc.

This difficulty has been overcome by the two methods.

A. Selective systems: have been designed, which facilitate the selection of mutants against normal wild type, as in case of biochemical, resistance and conditional mutations, where under certain conditions only mutants will grow permitting selection of one mutant among even a million individuals. Most of the selective systems are used in microorganisms.

B. Induced mutations are used, when selective systems are not available and therefore, frequency of mutations needs to be increased artificially, to allow convenient identification and scoring of mutations.

Induced Mutations: Mutations can be artificially induced with the help of mutagenic agents, which can be broadly classified into two groups, (a) physical mutagens-mainly radiations and (b) chemical mutagens.

Physical mutagens-radiations: Physical mutagens, are radiations, although change in pH value (acidity) or temperature shocks may also induce mutations. Classification of radiations commonly used for inducing mutations is given in the following table.

Ionizing radiations will cause ionization and will force ejection of an electron from the atom it attacks. But the non-ionizing radiations like UV do not cause ionization, but cause excitation through energy transfer. Among ionizing radiations, more commonly X-rays, gamma rays, beta rays, and neutrons are used for inducing mutations. X-rays are produced in a X-rays machine when energy charged particles like cathode rays (electrons) impinge on a suitable target like tungsten.

Similarly gamma rays are produced when an unstable atomic nucleus like Cobalt-60 releases energy to gain stability. Beta rays (electrons) are produced from element like phosphorous and neutrons are produced in a nuclear reactor. Some of the sources of radiations and their importance in induction of mutations are given below in the table.

Some radiation sources and their importance in induction of mutations

Radiation source	Radiation	Energy(MeV) (max.)	Half life	Penetration	Use
1. X-ray Apparatus	X-rays	0.05-0.37	-----	Many cm	Acute
2. Cobalt-60	Gamma rays	1.17 & 1.35	5.25 yr.	Many cm	Acute & Chronic
3. Cesium-137	Gamma rays	0.67	30 yr.	Many cm	Acute & Chronic
4. Phosphorus	Beta rays	1.71	14.4 cm	several mm	Chronic
5. Nuclear reactor	neutrons	Upto several Mev	-----	Many cm	acute

The effects of mutagen depends on its wave length and penetrance which are inversely correlated. Lower the wave length, higher the penetrance. This is why, ionizing radiations having lower wave length (λ) have high penetrance, and are thus used as acute radiation does which means that the dose of radiation can be supplied quickly within a few minutes in one installment.

Different kinds of radiations and their characteristics

	Types of Radiations	Wave length (nm; nm $=10^{-9}$ m)	Ionizing density
1.	Non ionizing Ultravoilet (UV) rays	10-390	------
2.	Ionizing (a) Electromagenetic rays X-rays Gamma rays	 0.10-10 10^{-4} - 10^{-1}	 8-200/µ 8-200/µ
	(b) Corpuscular rays Beta rays(electrons) Protons(H-nuclei) Neutrons Alpha rays(He particles) Other heavy particles	 ----- ----- ----- ----- -----	 8-200/µ 8-200/µ 8-200/µ 500- 5000/ µ & higher

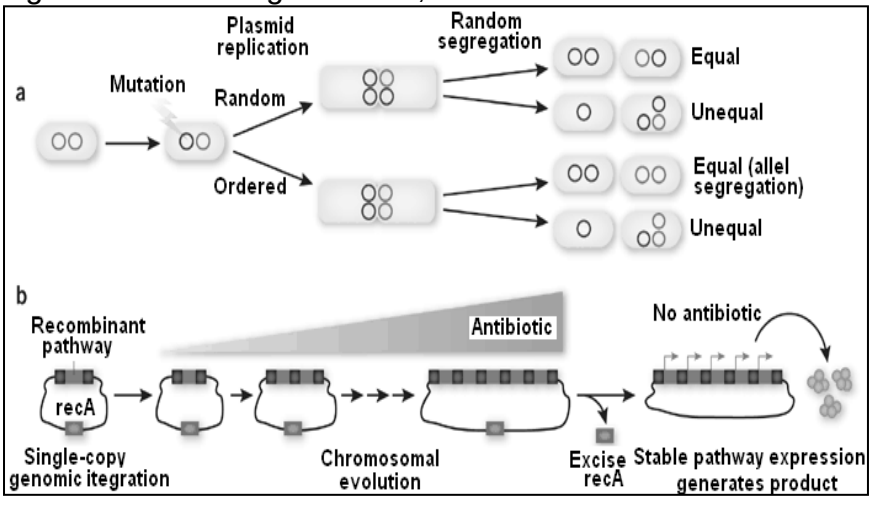
Such acute radiations are usually given to seed material. Contrary to this, some radiations are used in a chronic manner by keeping the living material like whole plants for seedlings in a gamma-garden where gamma rays are continuously emitted at a very slow rate. The mutagenic effect of X-rays and gamma rays are ionizing radiations and can induce

mutations in seeds. Ultraviolet rays, which are non-ionizing and have low penetrance are used for treatment of material where nuclei are not located too far away from the surface. Since the seeds, embryo is present several cell layers away from the surface of the seed; it is not suitable material for treatment with UV.

However, in plants pollen can be treated with UV, since every pollen has germinal nucleus, which can altered. Similarly, cells of bacteria and other microorganisms can be treated with UV rays, since many of them are unicellular or one celled thick.

Chemical Mutagens: Besides radiations, chemicals can also be used for inducing mutations. Mutations can also be induced due to certain chemicals. The chemical used for inducing mutations were mustard gas, ethyl urethane, phenol, formaldehyde, etc. mustard gas was found to be highly mutagenic & it was found to have a delayed effect.

For instance, if treatment is given in the first generation, mutations could be induced in second or third generation. A very long list of chemicals, which can be induce mutations, is now available. During the last two or three decades ethyl methane sulphonate (EMS) has been most extensively used for inducing mutations in microorganisms, higher plants and animals.



Chromosomal Aberration: A chromosomal aberration is an abnormality in the structure or number of chromosomes in an organism. Depending on the nature of the aberration, it can lead to severe birth defects, potentially including defects incompatible with life. Chromosomal aberrations can be diagnosed with the use of a karyotype, a visualization of an organism's complete set of chromosomes, where functional changes in the chromosomes will be readily visible. Treatment for people with chromosomal aberrations involves the provision of supportive care, as it is not possible to cure conditions caused by problems with the chromosomes.

The cause of a chromosomal aberration is usually a problem during cell division, either when cells divide to make gametes used in reproduction, or when cells are in the process of dividing in the embryonic state. Exposure to radioactive substances and chemicals known to cause mutations can cause abnormalities in the gametes that will be passed on to embryos in the event those gametes are used in fertilization. If errors occur during embryonic cell division, the resulting organism will exhibit genetic mosacism, where some of the cells have a healthy set of chromosomes, and others display a chromosomal aberration. Chromosome aberrations are classified as one of two types: numerical or structural. Structural changes involve the loss or gain of portions of chromosomes. The resulting patient may have "partial monosomy" ***/*** "partial trisomy."

Numerical abnormalities: Numerical changes are to two types: polyploidy with changes in the number of sets of chromosomes (polyploidy) and aneuploidy with changes in the number of individual chromosomes (e.g., trisomies and monosomies).

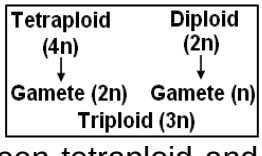
Introduction to polyploidy: Any organism in which the number of complete chromosome sets in higher than the diploid number is called polyploid. Terms described as follows:

1. **Aneuploidy**: The presence of chromosome number which is different than a multiple of the basic chromosome number.
2. **Euploidy**: The organisms should possess more than 2 sets of chromosomes e.g. shown in table.

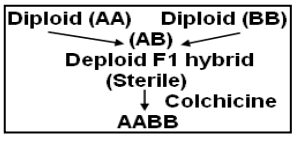
Sr. No	Number of chromosome sets	Types of polyploidy
1	One	Monoploid
2	Two	Diploid
3	Three	Triploid
4	Four	Tetraploid
5	Five	Pentaploid
6	Six	Hexaploid
7	Seven	Septaploid
8	Eight	Octaploid

Types of Polyploids: There are of 2 types:

a) Autopolyploids: These are the polyploids, which have the same basic set of chromosomes mutiploid. For e.g. if a diploid species has 2 similar sets of chromosomes or genomes (AA) an autoploid will have 3 similar genomes (AAA) and autotetraploid will bear genomes (AAAA). Triploids are formed by a cross between tetraploid and diploid plants. Diploids can be produced by fusion of 2 sperms with the same eggs. Triploids are usually sterile because the odd chromosome cannot undergo synapsis.



b) Allopolyploids: They develop due to hybridisation between two species followed by doubling of chromosomes. Allotetraploid (AABB) is the common example. Colchicine is used for artificially inducing polyploidy. Common cultivated



wheat is an interesting example of allopolyploid. There are three different chromosomes number in the genus Triticum namely 2n = 14 ; 2n = 28 ; 2n = 42. The common wheat is hexaploid with 2n and is derived from 3 diploid species.

Aneuploidy: refers to presence of abnormal number of chromosomes, and is a type of abnormality in chromosomes. Common cause of genetic disorders is presence of extra chromosome or absence of chromosome. Cancer cells also involve abnormal growth of cells or abnormal number of chromosomes. Aneuploidy mainly occurs during cell division when the chromosomes do not divide between the two cells. Chromosome abnormality occurrence is 1 of 160 live births. The most common extra chromosomes are 21, 18 and 13. Every cell in the human body has 23 pairs of chromosomes, a total of 46. One chromosome of each pair is inherited from the mother and the other chromosome is inherited from the father. The first 22 pairs of chromosomes, also called autosomes are numbered from 1 to 22, and are arranged in descending order in a karyotype. The last pair of chromosomes is called the sex chromosomes. Normal females have XX, while normal males have XY. At the time of meiosis, germ cells divide to create gametes, each gamete should have the same number of chromosomes. But sometimes, the whole pair of chromosomes will go in one gamete, and the other gamete will not receive any chromosome at all.

Structural abnormalities: Structural changes in chromosomes may be present naturally or can be artificially induced. There are four types of structural changes present in chromosomes:

1) Deficiency or deletion

- ✓ In case of deficiency, there is a loss of segment of the chromosome
- ✓ For pairing, the normal chromosome of homologous pair will form a loop (pachytene configuration)
- ✓ A very important example of deficiency is cat like cry in children which is associated with microcephaly (small head).
- ✓ That chromosome is now missing certain genes. When this chromosome is passed on to offspring the result is usually lethal due to missing genes.

2) Duplications or additions

- ✓ In case of duplication, there is addition of a part of the chromosome
- ✓ If the fragment joins the homologous chromosome, then that region is repeated.
- ✓ For normal pairing at pachytene, the duplicating chromosome of homologous pair will form a loop.
- ✓ Important example of duplication is bar eye character in Drosophilla, Fragile X syndrome, etc

3) Translocations

- ✓ Translocations may be defined as the process in which there is mutational exchange of chromosomal segments between non-homologous chromosomes. Or we can say a fragment of a chromosome is moved ("trans-located") from one chromosome to another
- ✓ Translocations are called as illegitimate crossing over
- ✓ For pairing the translocation heterozygote will form a plus shaped figure (pachytene configuration)

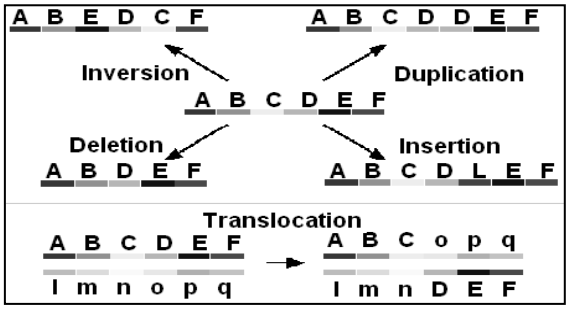
- ✓ Translocations were first observed in evening primrose and these were considered mutations by Hugo De Vries initially

4) Inversions

- ✓ In this case the chromosomes breaks at two points, the broken piece rotates through 1800 and thus reunites in reverse order.
- ✓ For normal pairing both the chromosomes of homologous pair will form loops

It is of two types:

- ➢ **Paracentric inversion**- If centromere or primary constriction is not present in inverted portion or segment, it is called paracentric inversion.
- ➢ **Pericentric inversion**- If inversion is present in inverted segment, it is called as pericentric inversion.



CHROMOSOMAL ABERRATIONS

In human beings various types of chromosomal variations, both numerical as well as morphological have been reported. Most of autosomal aberrations as follows

1,000,000 Conceptions

- 8,50,000 Live births
  - 8,33,000 Children
    - 5,165 Chromosomal abnormalities
      - 1,849 Sex Chromosome aneuploids (1247 males: 422 females)
      - 1183 Autosomal trisomies
        - 42 Trisomy-13
        - 100 Trisomy-18
        - 1,041 Trisomy-21
      - 758 Balanced Robertsonian translocation
      - 117 Inversions
      - 500 Unbalanced structural aberrations
  - 17,000 Perinatal deaths*
- 1,50,000 Sponataneous abortions
  - 75.000 Chromosomal abnormalities
    - 39,000 Trisonies (e.g., 3,510 Trisomy-21
    - 13,500 Turner syndrome(XO)
    - 12,750 Triploids
    - 4,500 Tetraploids
    - 5250 Others

**perinatal = pertaining to the period between 7th month of pregannacy & 1st week of life*

Occurance of various numerical and structural variations in human beings