UNIT I

CELL STRUCTURE & FUNCTIONS

1.1 CELLS ARE BASIC UNITS OF LIVING ORGANISMS

Cells are the smallest and the most basic structural and functional units of all living organisms capable of carrying on the activities of life. In other words, all living forms are composed of cells. Like the bricks of a building, cells are the "building blocks" of an organism.

Cells with similar functions can associate to form a tissue. Various types of tissues can function together to form organs. Organs then coordinate their activities to form a system, such as the digestive system or respiratory system. Finally, various systems together constitute an organism. Thus, all biological activities carried out by a living organism can be described in terms of cellular activities.

1.1.1 CELL THEORY

In 1838, Matthios Schleiden, a Dutch botanist, concluded that all plants are composed of cells. In 1839, the German zoologist Theodor Schwann published the work "Microscopic Investigation on the structure and Growth of Animals and Plants".

1. All organisms are composed of one or more cells
2. The cell is the structural unit of life.
   Rudolf Virchow, a German pathologist, completed the cell theory by proposing the third tenet of cell theory, which states that:
   Cells can arise only by division from a pre-existing cell.
3. Several more axioms have been added to these tenets to formulate the modern cell theory. The cell theory today states that:
   - **i.** The cell is the fundamental unit of structure and function in living beings.
   - **ii.** All known living beings are made up of cells.
   - **iii.** All cells arise from pre-existing cells by division
   - **iv.** Cells contain hereditary information (DNA), which is passed on from a mother cell to the daughter cells during cell division.
   - **v.** Some organisms are unicellular, that is, they are made up of only one cell.

vi. Other organisms are multicellular, that is they are made up of more than one cell (even millions of cells).
vii. Energy flow occurs within cells.
viii. All cells have basically similar chemical composition.

The following are the most important properties of cells:

- Cells are highly complex and organized structures.
- Cells can replicate themselves by binary fission, mitosis, or meiosis.
- Cells can acquire and utilize energy by the processes such as glycolysis and Krebs cycle.
- Cells can be motile and are capable of numerous mechanical activities.
- Cells respond to stimuli via cell surface receptors.
- Cells show osmoregulation, and can import and export substances to their surrounding through their membranes.
- Cell can evolve into different forms during the process of
- Cells die.

1.1.2 BACTERIAL CELLS

Bacteria are the simplest free-living organisms, that is, they can reproduce all by themselves and do not need a host to survive. Most of the bacteria grow on non-living surfaces. They are unicellular (single-celled) microorganisms found everywhere - soil, water, and air. Bacteria can live inside a human body but outside our body's cells. Most bacteria living inside a human body are beneficial to humans and help in processes like digestion, but some can cause infections. However, bacteria are responsive to antibiotics.

Bacteria were first observed by Antoni van Leeuwenhoek in 1676. He called them "animalcules". The name bacterium was introduced much later, in 1838 by Christian Gottfried Ehrenberg.

Bacteria range in size from approximately 10 to 100 times larger than viruses. Most bacterial cells are typically 0.5 - 5.0 µm (1µm= 10^{-9}m) in diameter. The smallest bacteria are about 100 to 200 nm (1 µm = 10^{-12}m) in diameter. Sizes of some common bacteria are shown below:

Shape	Arrangement	Examples
Coccus (Round shaped)	Monococcus Diplococcus Streptococcus(Chains) Staphylococcus(Bunches)	Micrococcus spp Streptococcus pneumoniae Steptococcus mutans Stapphylococcus aureus
Bacillus (Rod shaped)	Monobacilli Streptobacilli	Pseudomonas aureginosa Bacillus anthracis

	Irregular bacilli	Mycobacterium tuberculosis
Spirilla (Spiral shaped)	Single spirals	Treponema pallidium
Comma Shaped	Single cells	Vibrio cholera
Cresent shaped	Single cell	Selenomonas spp
Dumbbell shaped Rod	Single Cells in irregular	Corynebacterium diphtheriae
Pear shaped	Single cells	Pasteuria spp
Lobed spheres	Single cells	Sulfolobus
Pleomorphic	Single cells	Mycoplasma spp

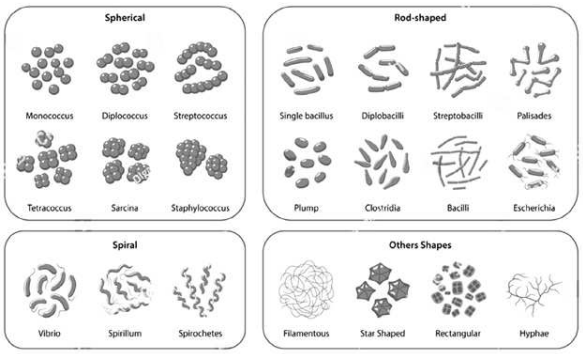


Fig. : Shapes of common bacteria

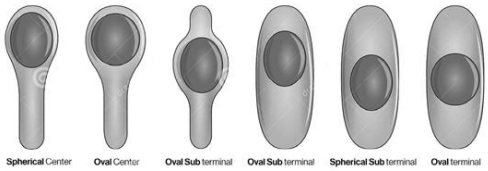


Fig. : Types of endospores

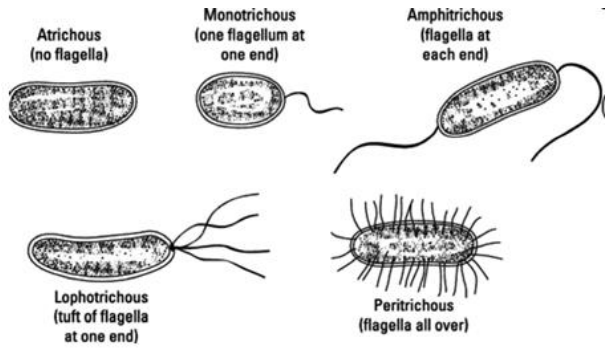


Fig. : Types of bacteria based on presence of flagella

Status of Flagella	Condition
No flagella	Atrichous
One flagellum	Monotrichous
Two flagella	Ditrichous
Many flagella	Multitrichous
A tuft of flagella at one end	Lophotrichous
A tuft of flagella at two ends	Amphitrichous
Flagella all around cell	Peritrichous

1.1.3 VIRUSES

A virus is much smaller than a cell. It needs to be within a cell (intracellular) to survive and derives its abilitity to multiply from its host cell. Viruses are very small (range in size about 25 to 300 nm), infectious obligate intracellular parasites. They can be isolated and crystallized almost like a chemical compound.

Viruses differ from living cells in the following aspects:

1. They either contain DNA or RNA, but not both.
2. Viruses do not divide outside of living cells.
3. Infected cells produce many copies of the virus particle.

☑ **Discovery of viruses:** Viruses were first discovered by Iwanoswsky in 1892. He termed them as filterable agents.

☑ **Morphology of Viruses:** The extracellular infectious virus particle is called the virion. A virion consists of a nucleic acid surrounded by a protein coat, called the capsid. The capsid with enclosed nucleic acid is known as the nucleocapsid. The capsid protects the nucleic acid. The capsid is composed of morphological units called capsomers. These are polypeptide molecules arranged symmetrically to form a covering around the nucleic acid core.

There are three kinds of symmetryshown by viruses - icosahedral, helical and complex (Figure)

- **Icosahedral:** The capsid is a polygon with 12 vertices (Corners) and 20 facets (Sides). Each facet is equilateral triangle. The capsid consists of 2 types of capsomers. They are:
  - ✓ **Pentagonal capsomers -** pentons on vertices, and
  - ✓ **Hexagonal capsomers -** hexons make up facets.

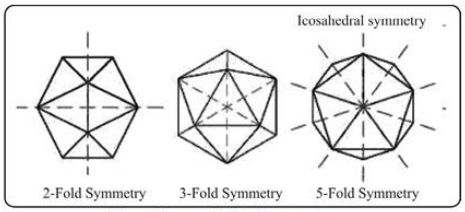
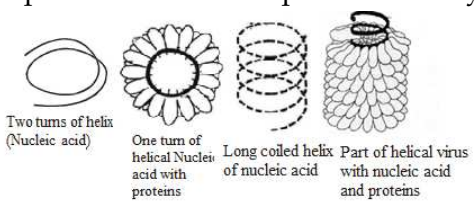
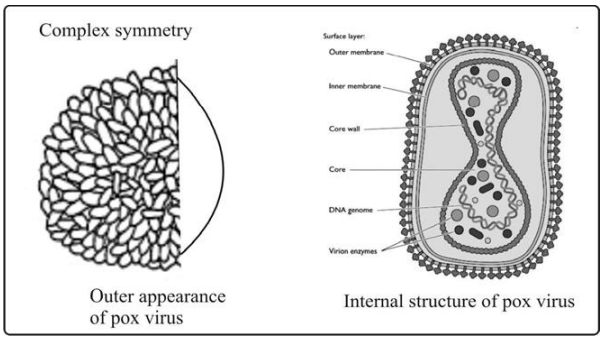


Fig. : Types of viral symmetry

- **Helical symmetry:** Capsomeres and nucleic acids are wound together to form a helical or spiral tube. An example of helical symmetry is TMV.



- **Complex symmetry:** Symmetry is not well-defined. Pox viruses exhibit complex symmetry. (Figure on next page). Viruses may be enveloped or non-enveloped. The viral envelope is derived from the host cell membrance. It is composed of……



Chemical properties of Viruses

- Viruses contain only one type of nucleic acid, DNA or RNA
- Viruses contain protein, which forms the capsid.
- Enveloped viruses contain lipids.
- Viral particles generally do not contain any enzymes, but retroviruses have a RNA-dependent DNA polymerase or reverse transcriptase.

1.1.4 FUNGAL CELLS

Fungi are simple eukaryotic organisms, which means that they contain nuclei. They represent very simple multicellular organization, where the body of the organism contains several nuclei but these may not necessarily be partitioned into individual cells. Fungi are free-living, but they do not have chlorophyll. A fungus is thus a saprohytic organism. It can survive independently and does not need a host to survive. A fungus can reproduce by sexual or asexual (vegetative) means.

Some fungi, like yeast, are unicellular. Other fungi are composed of microscopic filaments call hyphae (Singular - hypha). A hypha is ussually a tubular cell which is surrounded by a rigid, chitin-containing cell wall. Inside the outer tube-like wall, is the plasmalemma, a bilayered membrane, which surrounds the protoplasm. The hyphae grow by apical growth (extension of tips) to form a network to comprise a colony called the mycelium (Plural-mycelia). The hypae may be branched or unbranched. The hyphal wall consists of microfibrilas made up of hemicelluloses or chitin. True cellulose occurs only in cell walls of lower fungi. Hyphae occur in three forms:

1. Non-septate or coenocytic;
2. Septate with uninucleate cells; and
3. Septate with multinucleate cells

Hyphae contain nuclei, mitochondria, ribosomes, golgi and membrane-bound vesicles within a plasma- membrane bound cytoplasma. The sub-cellular structures are supported and organized micro-tubules and endoplasmic reticulum.

Some common types of fungal cells are shown in Figure.

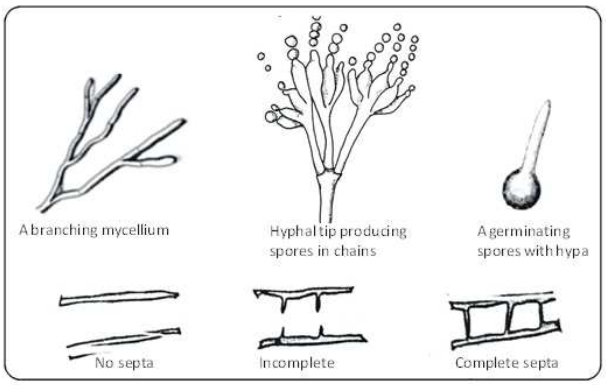


Fig. : Types of Fungal cells

1.1.5 PLANT CELLS

Plants are unique among the eukaryotes in that they can manufacture their own food. Chlorophyll, which is a green pigment enables plants to use sunlight to convert water and carbon dioxide in glucose, which then is utilized by the cell as fuel.

There are various kinds of plant cells. The most common types are:

1. **Parenchyma cells:** The simplest type of plant cell is called a parenchyma cell; most of the basic metabolic and reproductive processes of the plant occur in these cells. Parenchyma cells are usually depicted as the "Typical" plant cell because they are not very specialized. These cells synthesize and store organic products in the plant. Most of the plant's metabolism takes place in these cells.
2. **Collenchyma cells:** Collenchyma cells have a support function in plants, particularly in young plants. Pectin and hemicellulose are the dominant constituents of collenchyma cell walls. These cells help to support plants while not restraining growth due to their lack of secondary walls and the absence of a hardening agent in their primary walls.
3. **Sclerenchyma cells:** Sclerenchyma cells also have a support function in plants but unlike collenchyma cells, they have a hardening agent and are much more rigid. They are lignified dead cells forming fibers for increased support.

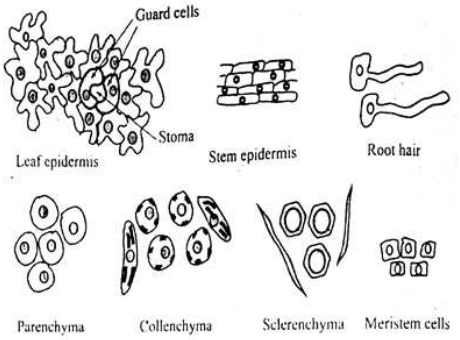


Fig. : Types of plant cells

1.1.6 ANIMAL CELLS

Animal cells have a structure typical of a eukaryotic cell, with the cytoplasm enclosed by a plasma membrane and containing membrane-bound nucleus and organelles. Unlike the cells of the two other eukaryotic kingdoms, plants and fungi, animal cells don't have cell wall.

The lack of a rigid cell wall appears to have allowed animals to develop a greater diversity of cell types, tissues and organs. Specialized cells that formed nerves and muscles - tissues not observed in plants – gave these organisms mobility. The ability to move about by the use of specialized muscle tissues is the hallmark of the animal world. (Protozoans also show locomotion, but by non-muscular means i.e. cilia, flagella, pseudopodia.)

Like plant cells, animal cells also have many organelles that perform various functions important to the cell's survival. Common types of animal cells are shown in Figure 1.6. Cells like red blood cells have to nuclei but straited muscle cells have many nuclei. Smooth muscle cells have a single nucleus each. The shapes of the nuclei are different in different white blood cells. Cells such as sperms have flagella while certain epithelial cells have cilia. Neurons, or nerve cells, have a unique structure with a long 'tail' covered by a myelin sheath. The structure of each cell is uniquely suited for its function.

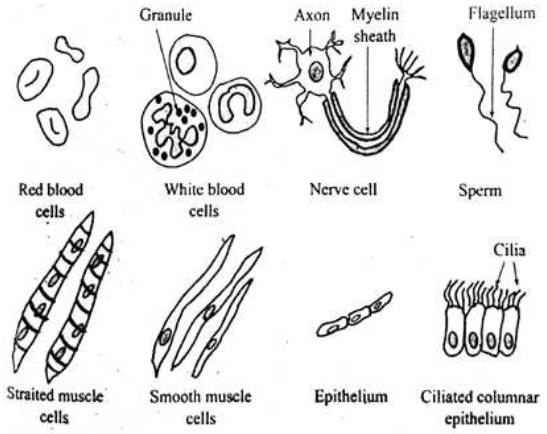


Fig.: Types of animal cells

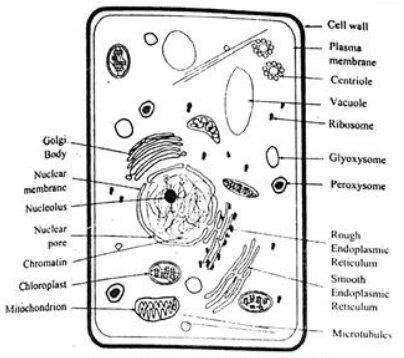


Fig.: Structure of plant cell

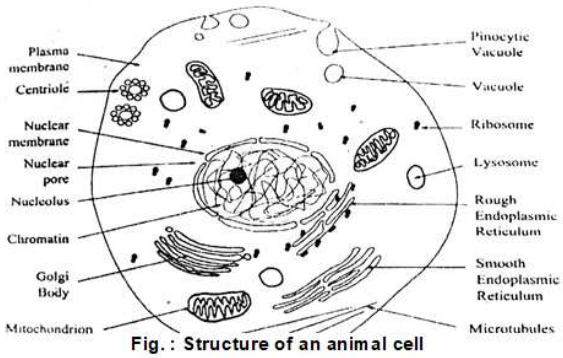


Fig.: Structure of an animal cell

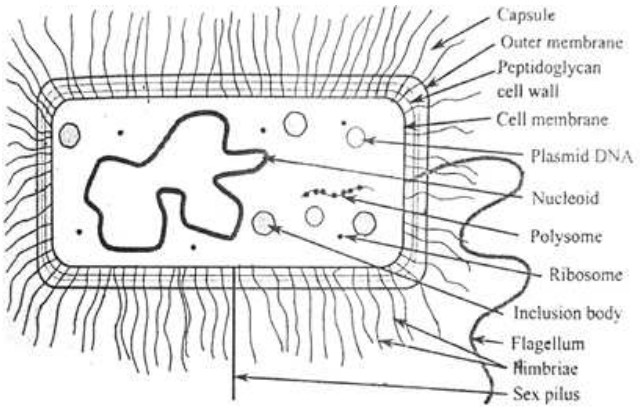
1.2 ULTRA STRUCTURE OF PROKARYOTIC CELL

A cell is fundamental unit of all living matter. As viewed under microscope, cells can be classified into two basic types based on whether or not a nucleus is present. Cells lacking nucleus are considered simple and primitive, and hence terms prokaryotic (karyon means nucleus). In contrast, cells that have a nucleus are considered complex and evolved, and termed eukaryotic.

Prokaryotes are diverse in both size and shapes. They vary in size ranging from 0.1-0.2µm to 50µm. The smallest prokaryote is PPLO -'Pleuro Pneumonia like Organism', belonging to the Mycoplasma group whereas the largest prokaryote is Thiomargarita namibiensis. A prokaryotic cell is the unit of structure of prokaryotes, which includes Archaebacteria, Cyanobacteria and Eubacteria.

A typical prokaryotic cell is bound by a rigid cell wall enclosing a cell membrane that surrounds the cytoplasm. The cell wall may be covered by sheaths, slimy layers, or capsules. In addition, several appendages may be present external to the cell wall: these structures include

(i) sheath
(ii) flagella
(iii) pili
(iv) fimbriae
(v) axial filaments.



The cytoplasm contains sub-cellular structures like

(i) nucleoid
(ii) ribosomes
(iii) plasmids
(iv) inclusion bodies
(v) molecular chaperones
(vi) thylakoids

1.2.1 PLASMA MEMBRANE

The plasma membrane is composed of phospholipids which form into lipid bilayers because they have a hydrophilic and a hydrophobic end. The hydrophobic ends align with each other to exclude water molecules. The hydrophilic ends face the interior aqueous environment of the cytoplasm and the periplasmic space or exterior of the cells. The functions of the plasma membrane of prokaryotes and eukaryotes are alike. A number of proteins are embedded in the lipid bilayer, which are primarily responsible for transport of ions, nutrients and waste across the membrane.

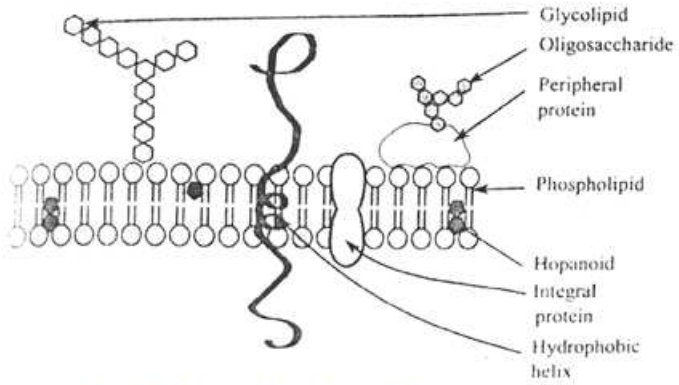


Fig. : Structure bacterial cell membrane

The fluid-mosaic model describes plasma membrane as a mosaic of both lipids and proteins.

1. Embedded or integral proteins that transverse all through the bilayer (and which can be removed only be destruction of membrane), as well as
2. Peripheral or surface proteins that are associated with only one face of the lipid bilayer (and which can be removed by mild treatments). The peripheral proteins are lipoproteins with lipid tails toward interior that help in anchorage and are hence also called lipid-anchored membrane proteins.

Functions of Cell membrane

1. Specific proteins in membrane allow passage of small molecule this is controlled by the membrane transport system.
2. The cell membrane has enzymes that participate in respiratry metabolism, and in the synthesis of capsular and cell components (peptidoglycan synthesis).
3. It is the site of generation of proton motive force - the force drives ATP synthesis and also causes flagellar motion.
4. Proteins/enzymes synthesized in cells are exported out the cell membrane. Examples of secreted products are lipoprotein amylases and proteases.
5. The plasma membrane is site of attachment of chlorosomes chlorobrium species. These are ellipsoidal vesicles that con... photosynthetic pigments.
6. It is selectively permeable allowing for movement of some compounds and inhibiting the movement of others. Transport across the membrane is through membrane transport proteins. There are three classes of transport proteins that are differentiated based of their function.

1.2.2 PLASMIDS

In addition to the nucleoid, prokaryotic cells often contain extra-chromosomal DNA in the form plasmids. Plasmids are present depending on the type of bacterial cells that a plasmid can colonize, and the type of other plasmids that they can co-ixist with, plasmids can be classified as:

1. Compatible plasmids, if two different plasmids can stably exist individually or together in a cell.
2. Incompatible plasmids, if two different plasmids cannot stably exist in the same cell. Such plasmids have the similar replication and partitioning systems, and are thus considered to belong to the same incompatibility group.
3. Narrow host range plasmids, if they can exist in only a limited number of closely related bacteria.
4. Broad host range plasmids, of they can be transferred to and maintained in a large number of host species

Based on their copy number, plasmids can be classified as:

1. Relaxed or multi-copy plasmids, which are present in multiple copies in a cell. Such plasmids are distributed randomly to daughter cells when the host cell divides.
2. Stringent or low copy plasmids, which occur in only few copies per cell. Such plasmids divide only when the host cell divides and are distributed between the daughter cells by a precise mechanism.

Range of environments in which the cell can survive. Based on their encoded functions, plasmids can be classified into the following six main classes. It is important to note that a single plasmid can belong to more than one category; for example, it can be a F-as well as a R-plasmid.

1. Fertility plasmids or F plasmids, which allow the host bacterial cell to conjugate or mate with another bacterial cell by the formation of a tubular connection between the two cells. The plasmid DNA (Sometimes along with some bacterial DNA) can pass from the donor cell to the recipient cell via this conjugation tube.
2. Resistance plasmids or R plasmids, which contain genes that metabolize antibiotics or other toxic substances to harmless products and thus allow the host cell to grow in the presence of these chemicals (confer antibiotic resistance). The 'R' factors were first detected in Japan in strains of Shigella that were resistant to many of the antibiotics used to treat bacterial dysentery.
3. Col plasmids, which produce bacteriocins or bacterial toxins caled colicins. Colicins are produced by some strains of E.Coli and can kill other competing strains. Colicins differ in their mode of action or killing action: ColE1 affects membrane-bound oxidative phosphorylation and interferes with ATP production, whie $ColE_2$ prevents DNA synthetis.
4. Degradative plasmids, which encode enzymes that can metabolize synthetic chemicals or xenobiotics such as salicylic acid, naphthalene and octane.

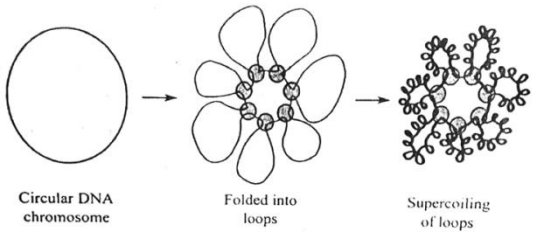
5. Metabolic plasmids, which encode enzymes for metabolic activities. For example, the Ti plasmid of Agrobacterium tumefaciens produces enzymes for the metabolism of opines. Other plasmids encode enzymes for denitrification activity in Alcaligenes spp., nitrogen fixation by Nif+ Rhizobium trifoli, and for utilization of simple compounds like sucrose, lactose, and urea.
6. Virulence plasmids, which produce toxins that can harm other livig beings and thus make the host bacterium a pathogen. For example, plasmids produce cfa (colonization factor antigen) in pathogenic E.coli, and enterotoxin in Staphylococcus aureus.

1.2.3 NUCLEOID

The nucleoid is the chromosome of a prokaryotic cell. It is also called the nuclear body or chromatin body and is placed centrally or eccentrically in the cell. In electron micrographs, the nuceoid in some cells appears to be associated with the inner face of the plasma membrane at one or more points. It is believed that such an association may facilitate separation of DNA into daughter cells after cell division. Note that prokaryotes do not have a membrane-bound nucleus, and the nucleoid is not membrane-enclosed.

Chemical analysis of nucleoids shows that the major component is DNA (~60%) which is complexed with RNA (~30%) and protein (~10%). The nucleoid can be observed under light microscope after Feulgen staining because the Feulgen stain binds specifically to DNA.

The loops are anchored to a central protein core. These proteins are similar to the histone proteins seen in eukaryotic chromosomes. One such protein is the Hu protein od E.Coli; Hu is a basic, heat-stable, DNA- binding protein. Another protein in E.coli that can bind DNA is the protein H. The amino acid composition of protein H is similar to that of histone H2A of eukaryotes. Other DNA binding proteins of prokaryotes are Hta from Thermoplasma acidophilus and H Bst from Bacillus stearothermophilus.



The major proteins involved in packing of DNA in E.coli are the 'histone-like' proteins HU, IHF, H-NS, and FIS. Each of these three proteins functions as a dimer. In addition to these proteins, small, highly positively charged compounds called polyamines, are also associated with the DNA. Examples of polyamines are spermine and spermidine.

1.3 ULTRA STRUCTURE OF EUKARYOTIC CELL

1.3.1 THE PLANT CELL WALL

1. **Middle lamella:** This is the first layer formed during cell divisions makes up the outer wall of the cell and is shared by adjacent celgluing them together. It is composed of pectic compounds and protein.
2. **Primary wall:** This is a thin, flexible and extensible layer formed after the middle lamella is formed. The cell may still grow after the primary wall has formed. Thus the walls of meristematic and parenchymal cells are composed mostly of primary cell walls. This layer consists of a rigid skeleton of cellulose microfibrils embedded in a gel-like matrix composed of pectic compounds, hemicellulose and glycoproteins.
3. **Secondary wall:** This is formed after the cell has stopped growing the secondary wall is extremely rigid and provides compression strength. It is made of cellulose, hemicellulose and lignin. The cellulose fibrils in this layer are deposited in alternating layers for additional strength. It contains pits or openings that make it fully permeable. The much thicker and stronger secondary wall accounts for most of the carbohydrate in the biomass of a plant. The secondary walls of xylem fibers, tracheids, and sclereids are further strengthened by the incorporation of lignin. Thus, the cell walls of sclerenchyma, collenchyma and xylem cells are predominant secondary cell walls.

The functions of the plant cell wall are to:

1. Provide structural and mechanical support to the cell.
2. Define and maintain the shape of the cell; cells are ultimately responsible for plant architecture and form.
3. Resist internal turgor pressure of cell.
4. Control the rate and direction of growth
5. Regular diffusion of material through the apoplast.
6. Store carbohydrates; in some seeds, the cell walls are metabolized.
7. Protect against pathogens, dehydration and other environmental fact.
8. Act as a source of biologically active signaling molecules.
9. Participate in cell-cell interactions

Plasmodesmata

Plasmodesmata are microscopic channels between the cytoplasms of adjacent plant cells; these provide direct connections or living bridges between cells. They are fairly abundant and densely concentrated in specific areas of the cell walls; comprising upto 1% of the cell wall area and present in up to one million plasmodesmata per square mm.

1.3.2 CELL MEMBRANE

It is approximately 7.5nm thick and forms boundary to the all and made up of Phospholipids (20-30%) & protein (60-70%).

Structure:

The plasma membrane is also called as cell membrane/cytoplasmic membrane.

- Singer .. Nicholson in 1972 the 'Fluid Mosaic Model' of plasma membrane. This model is well accepted.
- According to this model, the plasma membrane is Vivax-fluid structure, in which lipids and proteins are arranged in a mosaic manner.
- In plasma membrane, there are two types, they are also called as peripheral and Integral protein.
- The outer part of plasma membrane which is called as polar is mainted hydrophilic in nature whereas the internal part.
- Plasma membrane is reflexed the non-polar (induble in water) in which is maintained hydrophoble this condition is called as Amphiphatic.
- Hoponoids are st.. like substances which provides tability for plasma membrane permits the components and nutrients to move laterally.
- It is the site of genration of protein motive force. The force that drives ATP synthesis and also flagellax motion.
- It is selectively permeable allowing movement of some compounds and inhibiting the movement of other. Transport of compounds are by transport proteins. They are three classes.

1. **UNIPORTER:** Transport takes place from one side moment to other side (Only one direction).
2. **SYMPORTER (or) Co-porter:** Carrys additional molecules along with main molecules in the same directions.
3. **ANTIPORTER:** Transport of one molecule in one direction and other in opposite direction helps to maintain osmetic balance.

Function:

- The organic & inorganic nutrients are transported by permeases which are present in plasma membrane.
- It consists of enzymes required for biasynthesis pathway that synthesize components of all wall such as
- PGL, techoic acid, phospholipids LPS etc.
- It possess the attachment site for bacterial chromosome & plasmid DNA.
- The inner membrane invaginates to form meosomes as a site for Respiratory activities.
- It provides permeability barries & they helps in the absorption & escape to cell walls material outside the cell.

1.3.3 NUCLEUS

The nucleus is in the center of most cells and is generally the largest membrane-bound organelle in a eukaryotic cell. The nucleus is responsible for storing and transmitting genetic information. It is separated from the cytoplasm by the nuclear envelope which is composed of two concentric membranes joined at regular intervals to form circular openings called nuclear pores.

The pores allow RNA molecules and proteins to move through the pores and into the cytosol. Inside the nucleus, DNA and proteins associate to form a network of threads called chromatin. The chromatin is easily stained with basic dyes. Within the nucleus is darkly staining region called the nucleolus which is the site for the synthesis of ribosomal RNA.

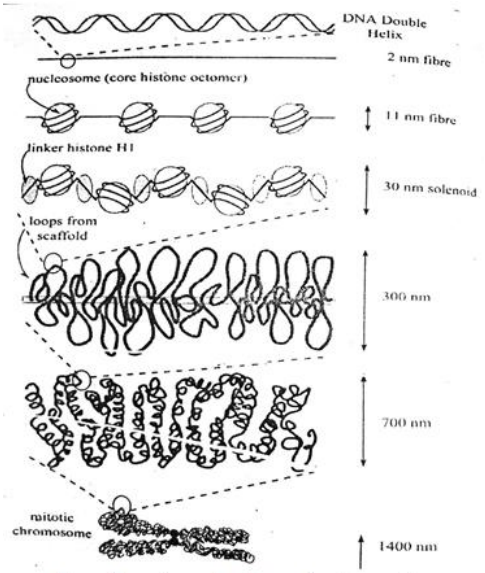


Fig. : Structure of eukaryotic chromatin

The fundamental unit of chromatin is the nucleosome. Each nucleosome is an octamer formed by the four core histone proteins. Two molecules of each of histones H2A, H2B, H3 and H4 associate to constitute the nucleosome, which is a bead-like particle ~11 nm in diameter. The DNA molecule, which has a diameter of ~2 nm, wraps around the nucleosome to produce a 'beads on a string' structure. Around 147 bp of DNA wraps around the nucleosome in around 1.6 turns. The stretch of DNA between two successive nucleosomes is called 'linker' DNA and this can vary from 10-18 bp.

1.3.4 MITOCHONDRIA

The mitochondria mean 'Thread granules'. They are filament in or granular cytoplasmic organelles of all aerobic cells of higher animals and plants and also of certain microorganisms including algae, protozoa, fungi they are absent in bacterial cells.

- **History:** Mitochondria was first do observed by Kollikes in 1850 as granular structures. Benda coined the term Mitochondria.
- **Distribution (or) Location:** Mitochondria has uniform distribution in cytoplasm.

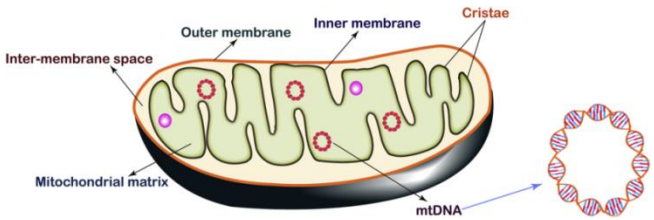
Morphology

1) **Number:** The number of mitochondria in a cell depends on the type and state of the cell. It varies from cell to cell and from species to species.
   Ex: The amoeba contain 50,000 & eggs of sea archin contains 1,40,000 to 1,50,000 etc.
2) **Shape:** The mitochondria may be filamentous or granular ... and may change from one form to other. Depending on the physiological conditions of the cells. Thus they may be sacket vesicular, ring or round shape.
3) **Size:** Normally mitochondria vary in size 0.5 m to 2.0 m some times their length may be reach upto 7µm.
4) **Structure:** Mitochondria have two membranes. The membranes are phosopholipid bilayer but inner and outer membrane are distincts in appearance and composition.

There are 5 components with in the mitochondria. They are:

1) **Outer membrane:** Outer membrane is composed of protein & phospholipids. It is composed of proteins & such as poreins which allows, free exchange of molecules (nutrients directly into the cytosol).
2) **Inter membrane space:** Inter membrane space is b/w outer & inner membrane filled with fluid.
3) **Inner membrane:** Inner membrane possess higher protein content. Which includes for the following purpose?
   Enzymes for
   - Oxidative phosphorylation
   - ATP synthesise
   - Specific transporter proteins

   Inner membrane is site for production of ATP.
4) **Cristae:** Inner membrane folds into number of foldings called cristae.
5) **Matrix:** The space formed by the inner membrane in foldings are called mitochondrial matrix. I contains enzymes required for several metabolites required for production of ATP it contains copies of ds DNA called Mitochondrial DNA/M..DNA.



Functions:

- Mitochondria perform most important functions such as oxidation, phosphorylation, dehydrogenation & respiratory chain of cell.
- They are the actual respiratory organs of the cell where the food stuffs i.e. carbohydrates and fats are completely oxidised into Co_2 & H_2O.
- During the biological oxidation of carbohydrates & fats large amount of energy is released which is utilised for the synthesis of energy rich compound known as ATP.
- As mitochondria synthesize large amount of ATP it is known as power house of cell.
- Besides ATP production mitochondria also serves as that production (or) thermiogenesis. Biosynthesis (or) anabolic activities and accumulation of Ca^{2+} & phosphate in animal.
- They also involves in Glycolysis, Krebs cycle, oxidative decorboxylation and respiratory chain & oxidative phosphorylation.
- It is also involved in process such as signaling cellular differentiation cell death , control cell cycle and cell growth.
- It is involved in several human diseases such as mental disorders, cardiac disfunctions & play a role in the ageing persons.

1.3.5 CHLOROPLAST

Chloro means green, plast means living.

During the process of photosynthesis chloroplast synthesis. The carbohydrates which contain energy in the form of chemical energy. The chemical energy is utilised by all living organisms to perform various life ac tivities. The chloroplast is green in colour due to the present of chlorophyll pigment.

- **Distribution:** The chloroplast are concentrated around the nucleus are just beneath the plasma membrane. Chloroplast are motile organelles & thus show passive & active (Chloroplast) moments.
- **Morphology:** Chloroplast are generally biconvex (or) plano convex in higher plants however varied from one plant cell to another that is it may be saucer shape, shperical, avoiv (or) clubshapes.
- **Size:** The size of chloroplast various from species to species & generally measure 2-3 µm in thickness & 5-10 µm in diameter.
- **Number:** The number of chloroplast various from cell to cell, species to species the cell of higher plants have 20-40 chloroplast.
- **Structure:** The chloroplast comprises of 3 main compartments.
  1) Envelope **2)** Stroma/matrix **3)** Thylakoids
- **Envelope:** The entire chloroplast is bounded by an envelope which is made up of double unit membrane, the envelope posses the layers they are
  1) Outer membrane **2)** Inner membrane **3)** Inter membrane space
  They contain 1-2% of total protein of chloroplast.

- **Stroma (or) Matrix:** It is also called as Matrix. It is maintained as a kind of jell, fluid phase surround's the thylakoid the stroma contain Rhibosomes & DNA which is involved in synthesis of structural proteins such as sugar, starch, fatty acid.

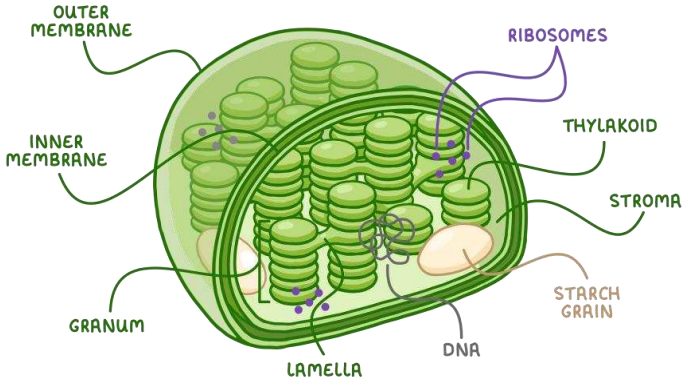
They contain 50% of total protein of chloroplast

- **Thylakoids:** Thylakoids are sack like membrane organelles which is stakk like a file of coins forming a structure called Grana. The numer of thylakoids per grana various fro m one to 50 or more. They form a network with in grana by lamella or intergranal fred. With in the thylakoids they are rich in photosynthetic pigments such as
  1) Chlorophyll a,
  2) Chlorophyll b,
  3) Carotenoids

  The remaining percentage of total protein comprises as Thylakoids

Function of chloroplast:

- It helps in the process of photosynthesis.
- They contain enzymes and co-enzymes necessary for photosynthesis. This is the site where light energy gets converted to chemical energy.
- It helps in electron transport system/chain by providing energy to move down or continue the energities.
- Palisade cells of leaves posses large numbers of chloroplast to increase the rate of photosynthesis.
- Chlorophyll pigment within the thylakoids of chloroplast helps in the absorption of light energy to continue Photosynthesis.



Structure:

Plastids are large cytoplasmic organelles found cell. They originated from colourless bodies called Photo Plastids They are 3 types of plastids.

1. **Leucoplast :** Colourless plastids found in roots.
2. **Chromoplast :** Colourless plastids found in flowers & fruits (Red, yellow or organe)
3. **Chloroplast :** Essential for photosynthesis & present pallaside cells of cells.

4. **Vacuoles :** Vacuoles are bubbles of material in the cell. Vacuoles hold water and can also hold solid materials and hold solutions.
   Vacuoles are bounded by single membrane called as Vacuoles membrane (or) Tonoplast.
   Young plant cells contains small vacuoles but as they mature all vacuoles unite together to form gaint central vacuole.
   The material seen in the vacuole is called as Sap. Cell sap contain salts, sugars, protein etc.

Function:

- It stores food
- It stores waste
- It stores ions such as ca, Na, Fe
- It helps in breakdown of macromolecules
- It involved in removal of waste products
- It maintains cells in internal water pressure

1.3.6 Endoplasmic Reticulum

The cytoplasmic matrix is transverse complex networks made up of inter connecting membrane bound vacuoles called as endoplasmic Reticulum.

- **History:** The name E.R. was coined in 1953 by portor.
- **Occurence:** The occurence of E.R. varies from cell to cell, it usually occupies surrounding the nuclear membrane area. It is also called as cytoplasmic vacuolar system (or) cytocavity network.

Morphology: They exist in three forms :

1) Lamellar form (or) Cisternae 2) Vesicular form (or) Vesicle
3) Tubular form (or) Tubules

Structure of E.R

- The cisternae, vesicles and tubules of endoplasmic Reticulum are made up of double unit mebrane (or) Phospholipid bilayer.
- The structure of E.R. continues with the membrane of plasma membrane, nuclear membrane, golgi complex.

Types of E.R

- There are two types of E.R.
  1) Smooth E.R. (or) Agranular 2) Rough E.R. (or) granular

Smooth E.R:

- This type of E.R. possesses smooth wall because of absence of Robosome the smooth E.R. abundant in cells which involves in the metabolism of lipids & steroids.
- The muscles cells are also rich in smooth E.R. which is called sarcoplasmic Reticulum

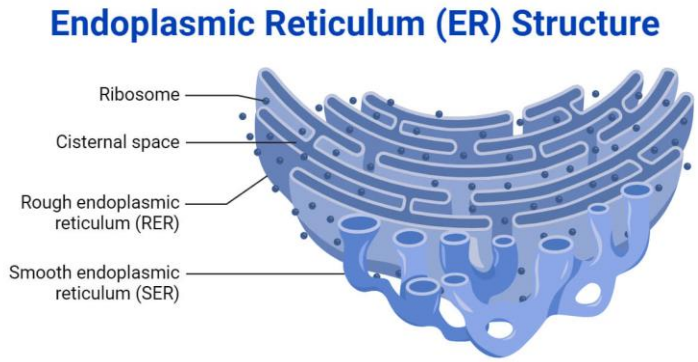
Rough E.R:

- The rough E.R possess rough walls due to the presence of Ribosome the cells which are active in protein synthesis possess rough ER.
- The association & dissociation of Ribosomes (603-603+405) occurs in Rough E.R

Functions:

It provides mechanical support to the cell internally by forming ultrastructural skeletal frame work.

- It involves in transport of proteins.
- It acts as supportive structure for the attachment of other organelle.
- Site for attachment of ribosomes for photosynthesis.
- It provides space for glycoprotein & folding of protein.
- Site for protection and storage of glycogen, steroids and other macro molecultes. They carry digestive enzymes for exocytosis.
- It acts as exchange of molecules by osmosis/diffusion.

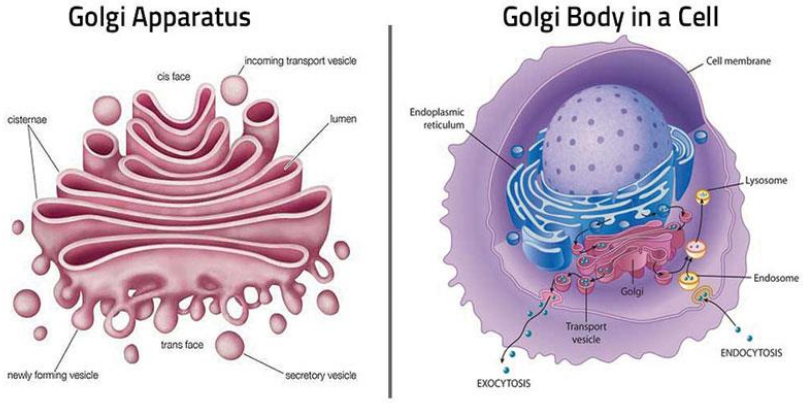


1.3.7 GOLGI COMPLEX

Golgi complex or golgi apparatus involves in important cellular functions such as biosynthesis of proteins, polysaccharides production of exocytic vessels and differentiation of cellular membrane.

- **History:** In 1898 Camillo golgi discovered Golgi apparatus for the first time. Due to their network like structure they are also known as Dictyosomes.
- **Occurance:** Golgi apparatus occurs in all cells except prokarydix and Eukaryotic cells of certain fungi.
- In animal cells, only single Golgi apparatus is present. Its number may vary from animal to animal and cell to cell.
- **Distribution:** In cells of higher plants, the golgi bodies (or) dctyosomes are usually found scattered through out the cytoplasm.
- **Morphology:** It is a membranous organelle composed of stackes of flattened vesicles or membrane sac called Cisternae.
- **Cisternae** are connected by complex flat tubules.

- Golgi body has 3-7 cisternae stacked one above the other file (or) plates. the stacks of cisternae has polarity. The two ends are called Cis-golgi and trane-golgi they two differ from thickness enzyme content & vesicle formation.
- The cis-golgi are associated with endoplasmic reticulum & receive vesicles containing protein.
- The protein reaches the lumen of cis-golgi through tubular conn the proteins then reaches trans-golgi. During this process the protein gets matured.
- The protein after maturing then buds off from the trans-golgi is vesicles, their vesicles are called smooth vesicles.



Functions of Golgi Apparatus

- ✓ Main function is of distribution and shipping for the cells chemical products. Synthesis and secretion of polysaccharides
- ✓ Formation of plant cell wall.
- ✓ Formation of lysosome and vacuoles Regulation of fluid balance of the cell Synthesis of cellular components
- ✓ Transfer of secretory proteins the cisternae of Rough endoplasmic reticu....
- ✓ Extra cellular transport of proteins to various organelles.
- ✓ Protein maturing occurs in the golgi body.
- ✓ Golgi complex is a center for reception, finishing, packaging & dispute for a variety of maerials in animals & plant cells.
- ✓ In plants, golgi apparatus is mainly involved in the secretion of most of primary & secondary cell walls. During cytokiness' of mitosis meiosis golgi apparatus is used in the formation of semi-solid is called cell plate.

1.4 FLUID MOSAIC MODEL OF CELL MEMBRANE

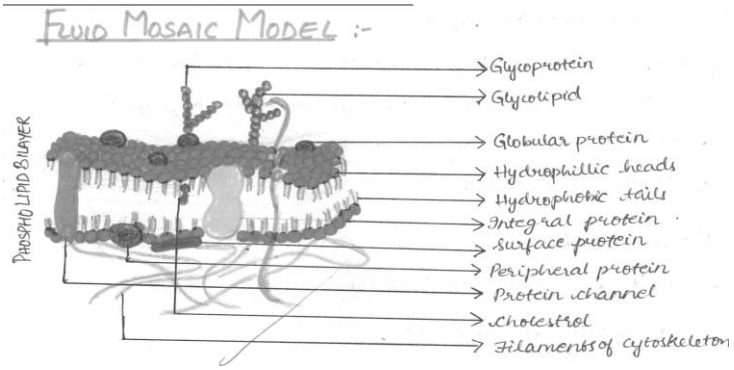
- ➢ 1st proposed by S.J.Singer and Gorth.L.Nicolson in 1972.
- ➢ This model describes plasma membrane as Mosaic of components such as
  - Carbohydrates
  - Phospholipids

- cholesterol
- Proteins

➢ These components gives Mosaic Characteristics to plasma membrane The ratio of these components vary with in cells Such as

Myelin contains 18% protein
76% lipids

Mitochondrial inner membrane contains
76% proteins
24% lipids



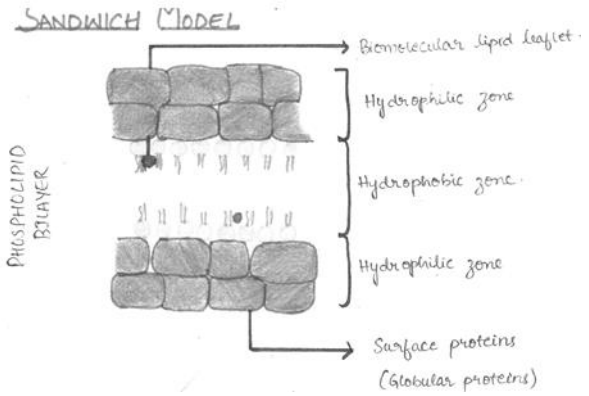
SANDWICH MODEL OF CELL MEMBRANE

☑ Davson-Danielli model proposed in 1935 by Hugh Davson and James danielli. This model also called as "Lipo-protein Sandwich".

☑ This model describes a phospholipid bilayer that lies between two layers of globular proteins and it is trilaminar and lipoproteinous.

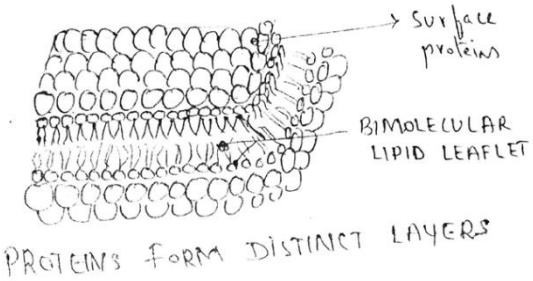
☑ Two layers of protein flanked a central phospholipid bilayer.

☑ Davson and Danielli model was predominated model until singer interact and Nicolson advanced the fluid Mosaic model in 1972.



☑ Sandwich model for plasma membrane structure is proposed model in which lipid bilayer is coated on either sides with Hydrated globular proteins.

☑ Hence plasma membrane is composed of two lipid protein bilayers. One facing the interior of the cell & other facing external to cells.



- ☑ Bimolecular lipid leaflet & surface globular proteins interact by Electrostatic interactions or vander walls bonds.
- ☑ Therefore polar ends of lipid molecule and charged amino acid of surface protein interact.
- ☑ Plasma membrane exhibit selective permeability i.e. it is able to distinguish
  - Molecules of different sizes
  - Solability properties
  - Ions of different charge
- ☑ Lipid bilayer is about 6.0 km & protein layer of about 1 nm. Total of 8 nm thickness.
- ☑ In Electron Microscopy, protein layer appeared dark & lipid bilayer appeared light colored region.
- ☑ Eukaryotic plasma membrane is lipo-protein in composition is per fludi mosaic model. The plasma membrane is made up of phospholipid bilayer. They are amphiphilic i.e. Hydrophilic are lipophilic (Fat- loving).
  Two types of proteins are associated with phospholipid bilayer. They are:
  1) **Extrinsic (or) Peripharal proteins -** They are bound with electrostatic & H2 bond Interactions.
  2) **Intrinsic (or) Integral proteins -** They internal and bond electrons Hydrophatic interractions.

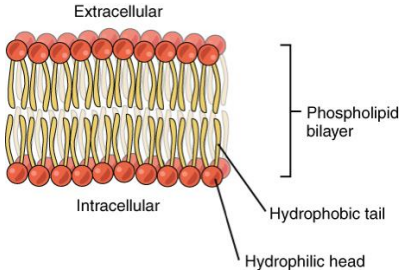
They belong to 3 categories:

1) **Marker Protein**
2) **Transport Protein** All float in the lipid layer and move freely
3) **Receplor protein**

Functions:

- ☑ It controls the transit of materials into and from cytoplasm. It is permeable to solutes
- ☑ Plasma membrane actively forms vesicles to take fluids by pinocytosis and take solids by phagocytosis.
- ☑ Plasma membrane is hydrophobic, hence it builds up the concentrations gradients of H2O soluble molecules and ions.
- ☑ It involves in holding cellular material

- ☑ It regulates the movement of materials across the membrane. It provides many chemical reaction.

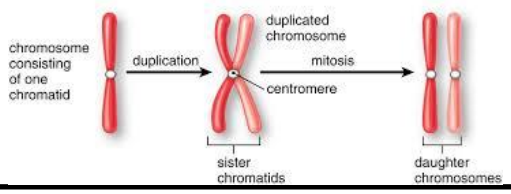


1.5 CHROMOSOME

- ☑ Karl Nageli (1842) first observed rid like structure present in nucleus of the plant cell.
- ☑ W.Waldeyes (1888) coined the term Chromosome.
- ☑ Walter Sutton and Theodor Boveri (1902) suggested that chromosome are pysical carrier of genes in Eukaryotic cell.
- ☑ Chromosomes are thread like structures present in the nucleus, which carries genetic information from one generation to another. They play vital role in
  - o Cell division
  - o Heredity
  - o Mutation
  - o Repair
  - o Regeneration
- ☑ In Eukaryotes, genetic material is present in nucleus in the chromosome which is highly organized DNA molecule with Histones supporting its structure.
- ☑ Chromosome number varies in different species. A human cell contains 23 pairs of chromosome. of which 22 are Autosomes & 1 sex chromosome.
- ☑ Karyotyping
- ☑ It is a technique to study the structure of chromosome present in a species. This technique is useful in finding out any chromosomal abnormalities.

Structure of Chromosome

Each cell has pair of each kind of chromosome c/a Homologous chromosome. Chromosomes are made up of Chromatin which contains DNA & Proteins. Each Chromosome contains 100s to 1000 genes. & Lode for many proeins structure of chromosome can be best seen during cell division.



Main parts of Chromosome:

i) **CHROMATIDS:** Each chromosome has two symmentrical structures called Chromatids which is visible in Mitotic Metaphase. But Anaphaze two sister chromatids gets separated & migrate to opposite poles.

ii) **CENTROMERE:** Sister Chromatid join at centromere. The position of Centromere differs in different chromosomes. This is also c/a primary construction. Centromere divide the chromosome into two parts.

- ☑ Short arm (or) 'p' arm
- ☑ Long arm (or) 'q' arm
- ☑ Centromere contains disc like structure called Kinetochore which has specific DNA seg.

iii) 20 Contriction & Nucleoler Organisers : This can be identified at Anaphase from centromere. Certain genes in 20 constriction form nucleds. These 20 constrictions are called as Nucleoler organisers (or) Figure. (NOR).

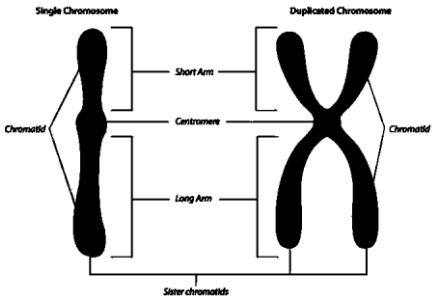
iv) **Telomere:** Terminal part of chromosomes is C/a Telomere. They are present at polar ends of Chromosome prevents the fusion of chromosomal segments.

v) **Satellite:** Elongated segment present on chromosome at 20 constrictions. These are called satellite & chromosome with satellite is called Sat-Chromosome.

vi) **Chromatin:** Chromsomes are made up of chromatin i.e it is made up of DNA, RNA proteins. At Interphase, chromatin fibres are present in nucleoplasm and chromosomes are visible.

After staining, darkly condensed region of chromatin is called Hetero-chromatin and contains Highly packed DNA which is genetically inactive.

Light stained region of chromatin is called Euchromatin and contains less condensed active DNA. At prophase, Chromosomal material is visible at thin filaments called as Chromonemata.



Q1.Write an Account of lamp brush Chromosomes?

Ans:

These are the largest known chromosomes found in the yolk rich oocytic nucleic of certain vertebrates such as fishes amphibians reptiles and birds. These are

discovered Ruckert (1892). They can rap be sean with the naked eye and are characterized by fine lateral loops, arising from their main axis, during first prophase of meoisis. These loops give it a brush like appearance. Hence are called lampbrush chromosomes. In certain urodele oocytes they may reach up to 5900 m in length.

Structure:

A lamp brush chromosome consists of 2 parts (i) main axis, and ii) lateral loops.

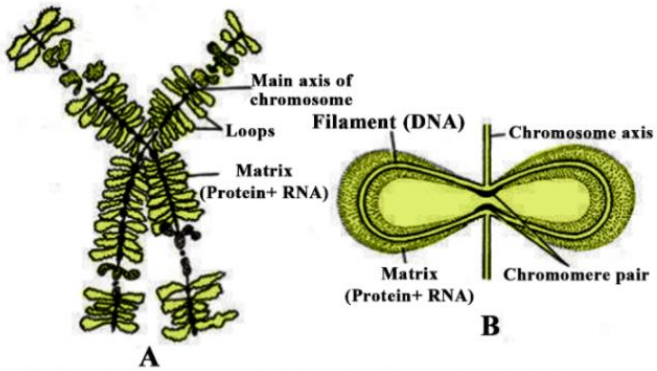


Fig. Lampbrush chromosome. A, Enlarge view of a part of lampbrush chromosome. B, One loop of a lampbrush chromosome

a) **Main axis:** It is the main line containing DNA and proteins and is continous with the axis of the loops. Thus loop axis is also composed of DNA and protein. It consists of four chromatids showing thick and thin insertion near the loop mouth.
b) **Lateral loops:** Main axis is composed of 4 chromalids (or) 2 bivalent chromosomes. The chromonema of these chromatids gives out lateral globular out grwoths, called loop axis. Around a loop axis is present matrixmade of RNA and proteins which gives it fuzzy appearance. At the base of each loop are darkly stained chromomers. The diameter of loop axis ranges from 30 to 50 $\frac{0}{A}$ Suggesting two double helixes.

These loops generally drop off or become reincorporated into the main axis of chromosome at the end of first prophase.

Near the chromomeres, matrix is thicker at one end, forming thicker insertion and thinner on the other side forming thinner insertion. Those loops may correspond to genetic loci.

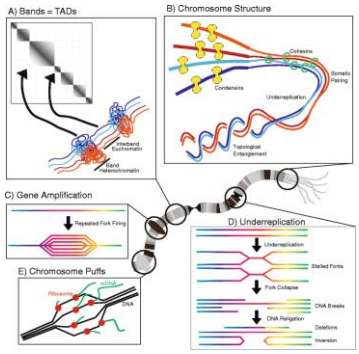
Functions:

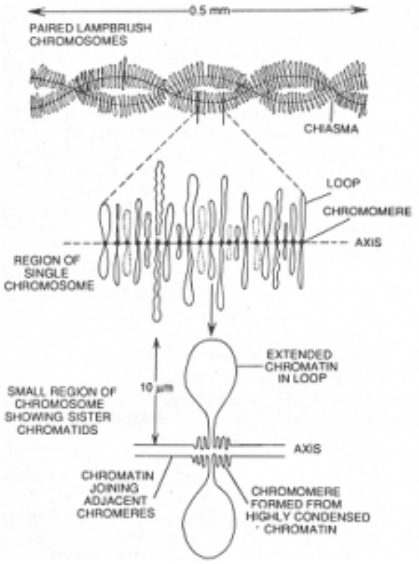
a) **Synthesis of RNA:** The functions of the lampbrush chromosomes involve synthesis of RNA and protein by their loops. RNA is synthesized only at the thin insertion and then carried around the loops to the thick insertion.
b) **Formation of Yolk Material:** Lamp brush chromosomes help in the formation of certain amount of Yolk material for the egg.

Q2. Polytene Chromosome

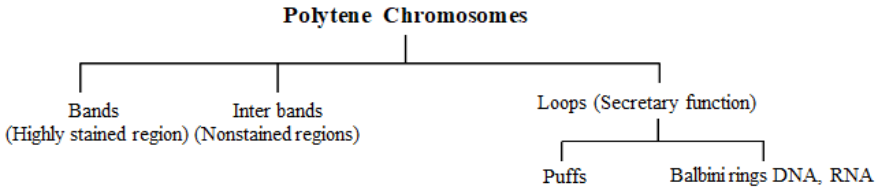
Ans:

Structure of polytene chromosomes: These are relatively smaller than the lamp brush chromosomes, found in the larvae of certain dipterans. In these larvae the Salivary glands contain salivary cells so large in size that they can easily be seen with the lens power of a dissecting microscope. The nuclei of these cells are much larger and the chromosomes in the nuclei are so large that they are 5 to 200 times as large.





1) **Structure**: Each polytene chromosome is a transversely straited structure consisting of a sequence of
   (a) dark-staining bands Separated **(b)** by non-staining interbands.
2) **Bands:** These are dark-staining regions composed of chromomers of individual chromonemata thus, the bands are due to a tigher coiling of the chromonemata in certain region than others. This banding was regarded as an expression of the linear sequence of genes in chromosomes. These bands are rich in DNA and small amount of RNA and basic proteins



a) **Interbands:** The non staining region situated between two successive bands is called an interband. Their length is variable.
b) **Puffs and Balbiani rings:** Bands of the chromosomes exhibit sweelings or puffs. The metabolic activities, required for the formation of puffs, are related to the secretary function of the salivary glands. This chromosomal RNA differes from the nucleolar and cytoplasmic RNA.

Some regions show large puffs than others, which are called balbians rings. These loops of chromonemata make up Balbiani rings and give the chromosome a fuzzy outlook. The Balbiani rings are rich in DNA, MRNA and are similar to the puff's in function and formation.

1.6 CHROMOSOMAL ABERRATIONS

The change in chromosome is due to alteration in genetic material through loss, gain (or) rearrangement of particular segments.

How does it happen

Non disjunction: Mistake in cell division where chromosomes do not separate in anaphace.

Polyploidy: Extra sets of chromosome but fatal in Humans

Anenploidy: Missing one copy or having Extra copy of single chromosome.

i) 3 copies of chromosome in somatic cell - Trisomy

ii) 1 copy of chromosome in somatic cell - Monosany Generally Autosomal Aneuploidy gets aborted

Anauploidy in humans sex Chromosomes

1. X-Female (Turners syndrome)
Occurs 1 in 2500 human females Short stature
Sterile (immatured sex organs) Reduced mental abilities

2. XXY male (Klinefelter syndrome)
Not detected until puberty
Female body characteristics develope Sterile
Reduced Mental abilities

3. XYY male (XYY Syodrome)
Very tall
Heavy acne
Aggressiveness
Mild mental Retardation fertile

4. XXX - female (Triple X syndrum)
More like XX females
Having 2 Barr bodies in sometime cell
Mostly sterile, if fertile, rises either XXY or XXX children

Aneuploidy in Human Autosomes

1. Autosomic monosomy: Fatal, usualy in Early pregnancy.

2. Autosomic Trisomy: Fatal, but individuals trisomic for autosomes at 13, 15, 18, 21 or 22 survive. Death can be observed before 1 yr of age.

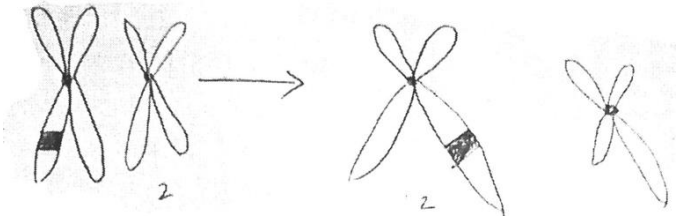
3. Trisomy 21 (Down syndrome)
Survival rate up to Adulthood

- ☑ Flat face, round head, epicanthi fold of eyes
- ☑ Adnormal facial appearance
- ☑ Mental retardation
- ☑ Develop Leukemia & Alzheimers Disease
- ☑ Hypotonia - poor muscle tone
- ☑ Short & broad hands

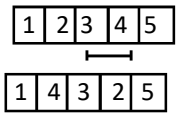
Abnormalities in Chromosomes ..... structure & rearrangements

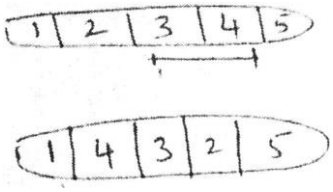
1. **RECIPROCALTRANSLOCATION**

A portion of 'Chromosome transferred to another.

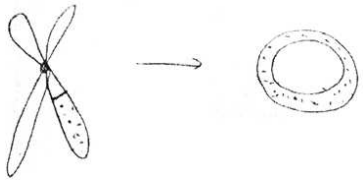


2. **INVERSION:** Inversion of portion of chromosome, broken off turns upside down & reattaches.

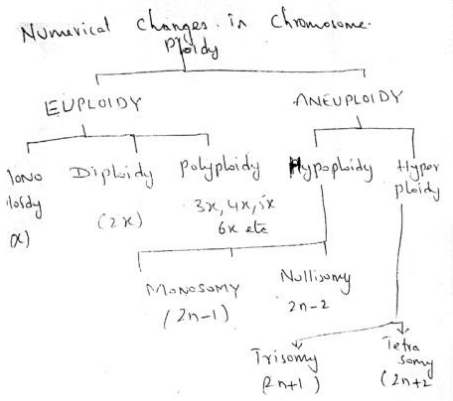




3. **DELETION:** A portion of chromosomes is missing.
   Eg: i) Wolf Hirschhorn syndrome, deletion in short arm of Ch-4
   ii) Jacobsen syndrome: Ch-11 'q' arm is deleted in term.....
4. **DUPLICATION:** Portion of chromosome is duplicated.
   i) Charcot-Marie tooth disease duplication of Myelin gene in Ch-17
5. **Rings:** Portion of chromosome segment broken off & form a circle or ring.



Numerical changes in Chromosome:



Fragile Sites:

Human X chromosome have regions that are poorly connected to rest of chromosome. Such regions are risk in CGG or CGC repeats i.e. is inherited like a gene. Breaks from these fragile sites leads to loss of Genetic material C/a FRAGILE X SYNDROME.

UNIT II

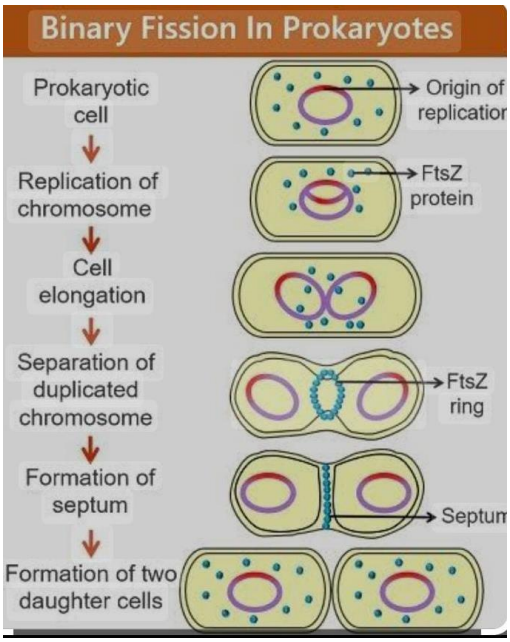
CELL DIVISION & CELL CYCLE

2.1 BACTERIAL CELL DIVISION

Asexual Reproduction – Binary fission

- Bacterial binary fission is a process by which bacteria carry out cell division. In eukaryotic cells, they divide by mitosis process.
- The Primary step in binary fission is to copy its DNA. The bacterial chromosome is found in specialized region of the cell called Nucleoid.
- Copying of DNA by replication process and involves many enzymes and begins at a spot on the chromosome called origin of replication.
- As replication continues, the 2 origins move at opposite ends of the cell, pulling the rest of the chromosome and thus separation of newly formed chromosomes.

Once, DNA moves into opposite cell end, division of cytoplasm takes place. In this process membrane pinches inward and a septum, a new dividing wall formed in the middle of cell.



Finally the septum splits and 2 cells are released and continues their lives as individual bacteria.

PROTEINS INVOLVED IN BINARY FISSION

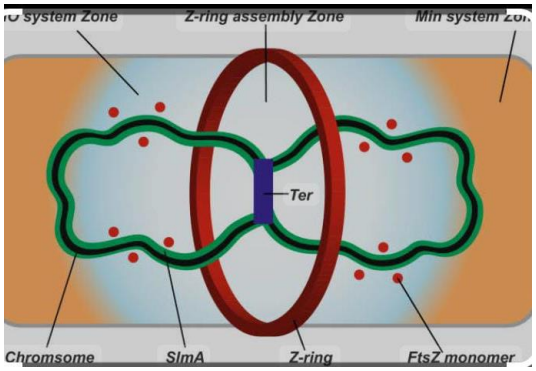
- FtsZ is 260 aa proteins involved in bacterial cell division and chromosome segregation.\ FtsZ is Filament Temperature Sensitive mutant Z. It is called Motor protein.
- FtsZ stabilizes septum and synthesizes in building membrane in septum formation.

CYTOKINESIS

- The process of septal invagination involves in growth of 3 layers of the cell envelope, they are

(i) Cytoplasmic membrane
(ii) Murein layer
(iii) outer membrane

- During cytokinesis, lines of FtsZ proteins would line up together parallel and poll on each creating "cords of many strings that tightens and separates the 2 cells apart.
- FtsZ binds to GTP acts as GTPase and excerts contractile force.



Steps

1. DNA is replicated
2. DNA attachment and cell growth
3. Pinching of cell membrane
4. Division of cells
5. Two daughter cells

Types of Binary Fission

1. Irregular
2. Longitudinal
3. Transverse
4. Oblique

2.2 CELL CYCLE

☑ The growing cell undergoes a cell cycle cells come from pre-existing cells doubles itself in six & then after a bried interval of time divides into two daughter cells. The daughter cell receives more or less half the material of the parents. This is a type of a sexual reproduction on Binary fission.

☑ In Eukaryotes cell divides by complex process called mitosis. The cell cycle can be described as a period of growth when nucleus & cytoplasm are growing life cycle of cell is divided into many stages. They are

1) Interphase
2) Prophase
3) Metaphase
4) Anaphase
5) Telophase

- ☑ All eukaryotes have to pass through Interphase to undergoe mitosis. The overall events of cell cycle are under the control of multiple gene products.
- ☑ The stage when the cell cycle is not dividing is called interphase.
- ☑ Interphase is sub-divided into 3 stages 1) G1 - phase 2) S-phase 3) G2 – phase

STAGES IN EUKARYOTIC CELL CYCLE

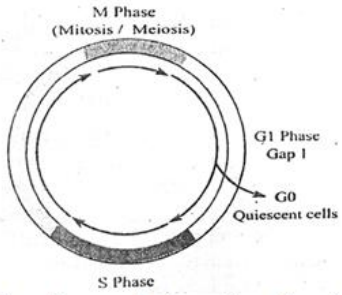


Fig. : Stages of eukaryotic cell cycle

M PHASE

- **G_1 Phase**
  - ☑ This is the first stage of interphase, the deration of G1 phase is highly variable. It may last for minutes (or) week, month (or) year.
  - ☑ Metabolic activity is minimum. The enzymes necessary fokr DNA synthesis during S-phase are synthesized during G1 - phase cell that never divide again such as Nerve cell are arrested in this stage.
  - ☑ Duration of arresting cell at G1 phase is at specific point. Such state is referred as Go state. Cells whichpass G - check point can proceed to S-phase.

- **'S'-Phase**:
  - ☑ It is also called as DNA synthesis.
  - ☑ In ths phase the DNA doubles i.e. DNA replication mechanism occur S-phase varies in duration. usually last for several hours. The S-phase is shorter, if DNA replication intiated at several location orisite, syntheiss of RNA and proteinsic lower expect histores which is required for packing okf DNA to form chromation.
  - ☑ The DNA content of Normal cell = 2c but by the end of S-phase the DNA content become = 4c.
- **G_2 - phase:**
  - ☑ It is also called as post DNA synthesis.
  - ☑ This phase is the period between end of S-phase & beginning of prophase. In this period synthesis of RNA & protein occurs for the formation of spindle fibres which are composed of microtubules. The duration varies but shorter than G_1 and S-phase usually last for two hours. After G_2 - phase the cell enter into m-phase & complete all stages

such as prophase, metaphase, Anaphase & telophase, after telophase the cell then enters, Interphase via G_1 , S_1 & G_2.

2.3 MITOSIS (SOMATIC CELL DIVISION) FOR REGENERATION OF SOME CELLS

Mitosis

- ☑ The term mitosis means thread in Green Mitosis cell division was first discovered by flemming in 1879.
- ☑ In Mitosis 2 chromatids of each chromosome separate and more to the opposite pole of cell. The two daughter cells look alike as parent cell. Somatic cell of all organisms divide by mitosis.

Stages of Mitosis:

- ☑ There are 2 stages of mitosis. a) Karyokinesis b) Cytokinesis
- ☑ 5 stages Inter phase protasemeta Ana Telophase

Prophase:

- ☑ Chromosomes becomes visible as this threads the entire prophase is divided three sub-stages.
  - **1)** Early prophase
  - **2)** Mid prophase
  - **3)** Late prophase

In prophase condensation of chromosomes occur inside the nucleus. In early prophase the chromosomes are evenly distributed in the nuclear matrix & in the midphase the chromosomes become shorter & thicker, & moves towards nuclear envelope, disintegrates & reduction of size of nuclei. In cytoplasm spindie fibres from antrioles. The sister chromatids of each chromosome are coiled to each other and referred as relational coiling.

- **a) Plectonic coiling:** Occurs between 2 sister chromatids they are turisted around each other can be separated when rotated.
- **b) Paraneomic coiling:** They are not tyrusted ariybd but remain attached. They cannot be separate when rotated.

Metaphase:

Complete dissolution of nuclear membrane & arrangement of all chromosomes on the equiatorial plate between the two spindle at the poles. Each chromosomes of both poles get attached by spindle fibres each chromotids made up of two sister chromatids. Each sister chromatids connects to spindle fibres by kinetochore a structure formed by centromere.

- ☑ The sister chromatids gets separated because of the absence of relation coiling.
- ☑ The main features of metaphase are 2 sister chromatides

- ☑ Absence of nucleoules.
- ☑ Disappearence of nuclear membrane.
- ☑ Arrangement of chromosomes on equaterial plate & called as Metakinesis, kinetodhors formation by centromere.
- ☑ Condensed state of chromosomes.
- ☑ Spindle fibres centrioles are formed.
- ☑ Centrioles present on both the poles.

Anaphase:

In this phase, the chromosal fibres of spindle contract leading to shortening of spindle fibres upto 1/3 rd of to 1/5th of its original size. This causes the chromosomes to split into two sister chromatids. Each chromatid is attached to spindle fibre through its kinetochore. The sister chromolids migrates towards oppsite poles. Based on positon of centromere there are stypes.

1) Meta centric chromosomes - V shape
2) Sub meta centric chromosomes - L shape
3) Telocentric chromosomes - Rod shpae

The spindle fibres which passes from one pole to another is called continuous fiber. The spindle fibkre passess from one pole to centromere of sister chromatid it is called chromosomal fiber.

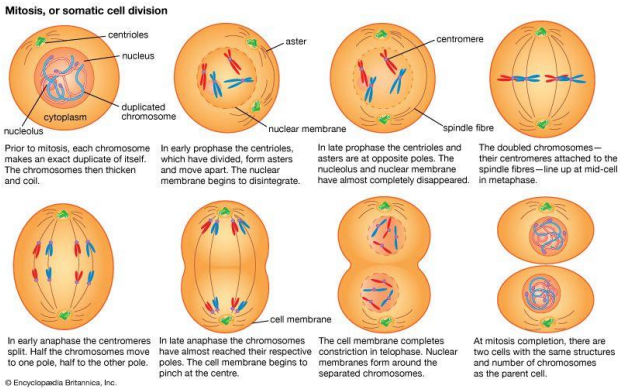
Meta centric Submeta centric Acrocentric

Telophase:

- ☑ The last stage of M-phase.
- ☑ The chromosomes decondence like prophase Nucleoli reapears.
- ☑ Nuclear envelope forms around each group of chromosomes Cytokinesis is intiated.

Cytokinesis:

- ☑ This stage is also called as cell clearage.
- ☑ The processes of formation of two daughter cells differ in animal and plant cell.
- ☑ In animal cell, the equatorial, contraction originates from outside the cell and moves inward till mid point forming microfibres and cell plate. This results in two daughter cells. Microfibers are made up of action & myosin.
- ☑ In plant cell it occurs by formation of phragm plast made up of microtubules and golgi vesicles which is made of protein. The tubules and vesicles fuse to form a primary cell wall of daughter cell.
- ☑ The fraction of dividing cells in a population of cells called mitotic Index (MI) high / MI population is expanding / low MI population is not dividing.



Significance of Mitosis:

- ☑ **Vegetative Growth of organisms:** Which leads to increase the size of entire organisms (or) selected organs tissues?
- ☑ **Cell Replacement & Regeneration:** This is the only route to replace the damaged (or) ageing cells via mitosis.
- ☑ **Asexual (or) Vegetative Reproduction:** It is a simple process by which plants & animals produce off spring which is genetically similar to parent cells.
- ☑ **Mitosis** makes it possible to grow tissue in culture such as animal and plant tissue culture to produce identical cells (or) clones.

2.4 MEIOSIS

- ☑ **GAMETOGENESIS**
- ☑ SEX CELLS — MALE (SPERM)
  FEMALE (EGG)
- ☑ INTERPHASE (4 PHASES)
  G_1 S G_2 M
- ☑ MEIOSIS TWO PARALLEL PHASES IN INTERPHASE THEY ARE MEIOSIS I & MEIOSIS II
- ☑ CYTOKINESIS
- ☑ PROPHASE-I, METAPHASE-I, ANAPHASE-I, TELOPHASE-I
  PROPHASE-II, METAPHASE - II, ANAPHASE-II, TELOPHASE-II
  KARYOKINESIS
- ☑ MEIOSIS-I (PROPHASE-I) DURING MEIOSIS

LEPTOTENE ZYGOTENE PACTYLENE DIPLOTHENE DIAKINESIS

MEIOSIS

1. Meiosis takes place during producing of gametes (as gametogenesis in sexually reproducing organisms.
2. In meiosis, mother cell divides to produce four daughter cell. Each with half of number of chromosomes as the mother cell.

3. The cell preparing to undergo meiosis also enters through G1, S, G2 phase which is just like mitosis but meiotic cell undergo 2 rounds of division called Meiosis I & Meiosis II to produce four nuclei and four cell.

Meiosis - I 1) Prophase-I
2) Metaphase-I
3) Anaphase-I
4) Telophase-I

Prophase Leptonema
Pachytema
Zygotema
Diplotema
Diakenesis

... phase-I

- ☑ This is the first stage of Meiosis
- ☑ **Leptonema stage:** Lepto = thin, nema = thread .
- ☑ There is increase in nuclear volume
- ☑ The chromosomes condensos & become visible as fine threads.
- ☑ Proteins required for chromosomal condensor are synthesized.
- ☑ Sister chromatids are not distinguished chromatids fibers comprises of chromosomes fine thread like structure called chromonema. Chromonema show thicker granule like structure called chromomere.
- ☑ **Zygotema:** Zygon = Adjoining
- ☑ Homologous chromosomes paired up through out its length.
- ☑ This is pairing up of homologous chromosomes is called as Synapsis
- ☑ After pairing they form a complex called Synaptonemal complex This complex is flat, attach to nuclear envelope at their telomeres.
- ☑ This complex is very essential for crossing over.
- ☑ **Pachytema:** (Pachus = thick)
- ☑ Further condensation of chromosomes & becomes shorter & thick Pairing up of homologous chromosomes is completed.
- ☑ The maternal and paternal chromosomes pair up and appears half the total number.
- ☑ Hence there chromosomes are called Bivalent (or) Fetred stage (or) four stard.
- ☑ Less % of DNA synthesis takes place.
- ☑ **Diplotema:** (Diplo-seperation of into two)
- ☑ In this stage, homologous chromosomes such bivalent gets separated.
- ☑ The separation is incomplex become homologous chromosemes remains in contact at the points of crossing over. These points are called 'Chiasmata'.
- ☑ The four chromatids becomes distinguishable and synaptonemal complex becomes invisible.
- ☑ This is a long lasting period. In human oocyte reach their stage by fifth month of factal development.

Diakinesis (dia-2cross)

- Chiasmata terminilization is completed.
- 2 homologous chromosomes of each bivalent are attached at (or) ... to one (or) both the telomeres only. Chromosomes further condenses & becomes shorter & thickness.
- Nucleolous & Nuclear envelope disappears.
- Spindle fibres are organized.
- The bivalents migrate to the equatorial plate of the cells.
- Bivalents are in 3 forms
  - **1)** Ring Bivalent
  - **2)** Open Ring Bivalent
  - **3)** Rod Bivalent
- Both the ends of meta centric and sub metacentric forms chiasma then it form closed ring bivalents.
- If one end of meta centric and sub-meta centric forms chiarma then it forms open ring bivalents.
- Acrocentric & Telocentric forms Rod bivalent.

METAPHASE-I:

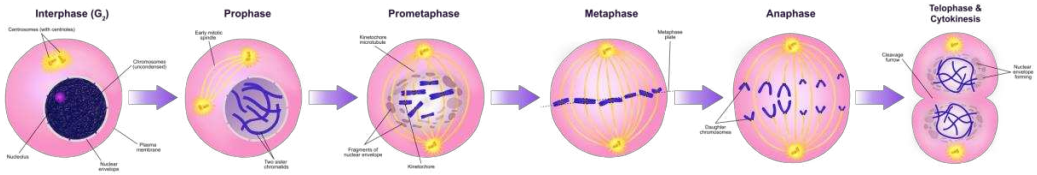
1) Nucleolus disappea
2) Nuclear membrane disintegration
3) Chromosomes arranged in equratorial plate
4) Spindle fibres are formed & attached to Kinetochores of sister chromatids.

ANAPHASE-I:

1) Seperation of two homologous chromosomes of each bivalent.
2) Migrate towards the poles spindle fibres disappears.
3) The number of chromosomes in the cell is half (n)

TELOPHASE-I:

1) Chromosomes uncoil
2) Nuclear envelope & Nucleolus becomes organized
3) This stage ends the Meiosis



MEIOSIS-II

Interphase-II - It is short resting stage b/w Meiosis I and Meiosis-II

Prophase-II -

1) Nuclear membrane
2) Chromosomes are condensed
3) Four spindles appratus are formed two of each of nearly formed.

Metaphase-II

1) Chromosomes paired up of equatorial (Right angles to Metaphase I)
2) Spindle fibres are attached to sister chromatids via Kinetochorm.

Anaphase-II - 2 Sister chromatids are pulled a part by shortening the lengh of spindles fibred.

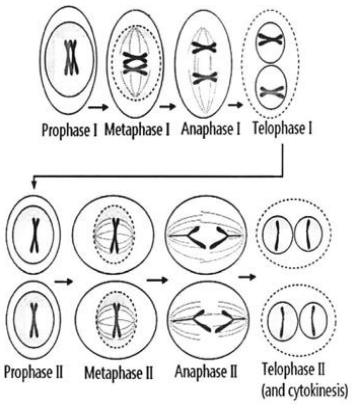
Telophase-II

- Chromatids complete migration & decondense
- Nuclear membrane forms around each of the four nuclear.
- Each of the four nuclei by the end of telophase-II one parent cell product 4 haploid daughter cells. These 4 daughter cells together is refe as Tetrad.

Significance of Meiosis:

- Production of haploid (n) gametes so that fertilization restores the normal somatic (2n) Chromosomes.
- Segreggation of 2 cells alletes of a gene due to pairing b/w 2ho chromosomes.
- Independent segreggation of alleles located in separate chromatins at metaphase-I
- Recombination b/w linked genes occurs during pachytone stage.
- Generation of genetic variation through segreggation, indep assortment & recombination.

Steps of Meiosis



Significance of Meiosis

1) Meiosis helps to maintain the constant number of chromosomes by reducing the chromosome number in gametes.
2) Meiosis is essential for sexual reproduction in higher animals and plants.
3) Meiosis helps in the formation of haploid gametes and spores for sexual reproduction.
4) Number of chromosomes in meiosis are fixed in a species for generation and generation.
5) Crossing over occuring brings the genetic variation in off-springs which helps in evolution of organisms.

6) The random distribution of maternal and paternal chromosome take place in to daughter cells. While meiosis and it is sort of independent assortment which leads to variation.
Meiosis: Process of Gametogenis gives 4 Non identical daughter cells.
Sex cells - If it is female (Egg) If it is in male (Sperm)

2.5 SENESCENCE AND NECROSIS

1. Stress response and DNA damage

Cells can reach senescene by various factors such as

- Shortening of Telomers
- Oxidative stress
- DNA damage response
- Double strand breaks
- DNA repair, etc.

Role of Telomers:

Telomers are DNA tandem repeats which are present at the end of the chromosome, which shortens during each cycle of cell division.

2.5.1. SENESCENCE

1. Cellular senescence is defined as a condition in which a cell no longer has ability to proliferate. These cells are Senescent cells that are arrested at G1 phase of cell cycle but they are mtabolically active.
2. Senescent cells undergo a phenomenon characterized by Cessation of cell division.
3. Leonard Hayflick in 1960's found that Human fetal fibroblasts becomes senescent after 50 cell population doubling. This is known as **Hayflick limit**.

Signalling Pathways

1. P^{53} pathway
2. P^{16} pathway

P^{53} pathway

P^{53} Activates P21

⇩

P21 deactivates cdk2 (Cyclin dependent kinase)

⇩

(PRB) Retinoblastoma protein remains active without cdk-2

⇩

PRB blinds to transcription factor

⇩

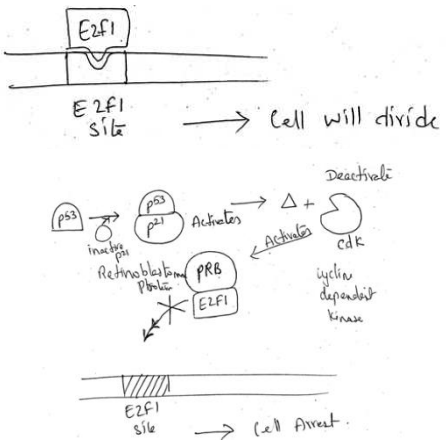
E2F1

⇩

This represses targets of E2F1

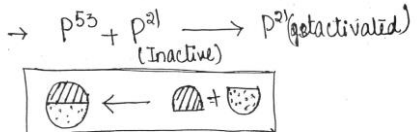
⇩

Positive

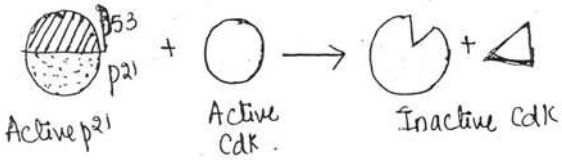


P53 Pathway

- It is important signalling Pathway in Senscent cells)
  P53 - Protein, 53
  Separated by S.D.S-Page
- P53+ P21→P21 (get activated).
  (Inactive)

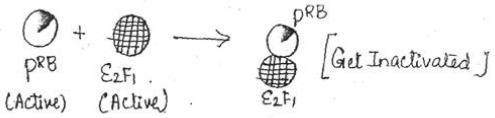


- P21 + Cdk Cdk →(Inactive)
  (Active) (Active)



- Cdk + PRB → PRB (Activated)
  (Inactive) (Inactive)

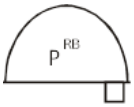
- PRB + E F → E F (Inactive)
  (Active) (Active)

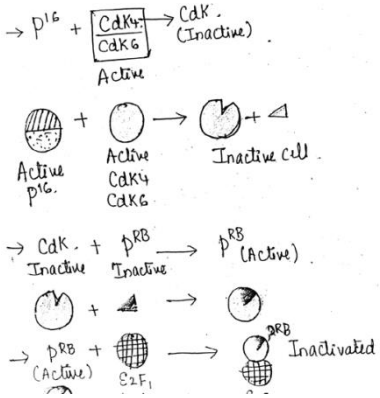


P53 Signalling pathway will make to move normal cell to senescent cell

- Cdk → Cyclin dependent kinen
  PRB → Rectino Blastoma protein

P16 PATHWAY





is actived by inactivating cdk4 & cdk6 by P16
(inactive state)➔ Active PRB binds to E F➔Cell arrest

Clearance of Senescent Cells

- Natural killer cells (NK)
- Macrophages
- NK cells directly kills senescent cells
- The NK cells also release cytokine which activates Macrophage & phagocytized the senescent cells.

2.5.2 NECROSIS

Necrosis in Greek means "death". It is a form of cell injury which results in premature death of cells. Cell Necrosis occurs by External factors such as

1) Infection
2) Trauma

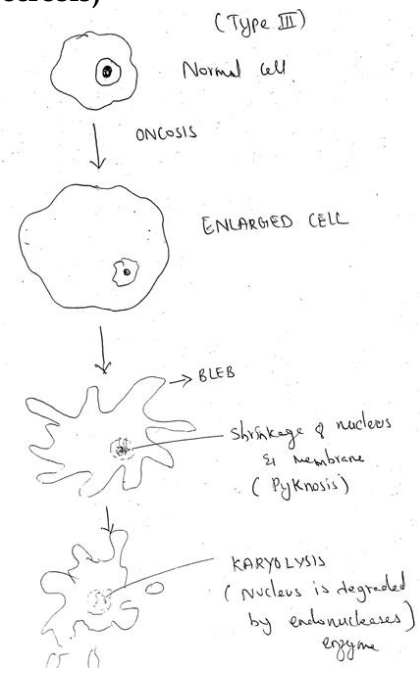
and also by Internal factors such as

1) NK cells
2) Macrophase

There are six types of patterns of Necrosis

1) **Coagulative Necrosis:** Under low oxygen environment (Hypoxic), there is formation of gel-like substance in the tissues. Due to denaturation of protein i.e. albumin converts into firm state.
2) **Liquefactive Necrosis:** Due to bacterial or fungal infection dead cells forms viscous liquid mass & in this liquid is yellow color with the presence of dead lenkocytes. This necrotic fluid is called Pus.
3) **Gangrenous Necrosis:** When coagulative necrosis continuous with liquefactive necrosis, this super imposed infection of tissues are called wet gangren.
4) **Caseous Necrosis:** Combination of coagulative & Liquifactive necrosis caused by Mycobacterum which appears white & friable (Cheese) Dead cells are not completely digested leaving granular particles.
5) **Fat Necrosis:** This results by the action of lipases on fat tissue. In pancrease Pancreatitis.
6) **Fibrinoid Necrosis:** Special form of necrosis when antigen binds with antibody which then deposits in arterial walls along with fibrin.

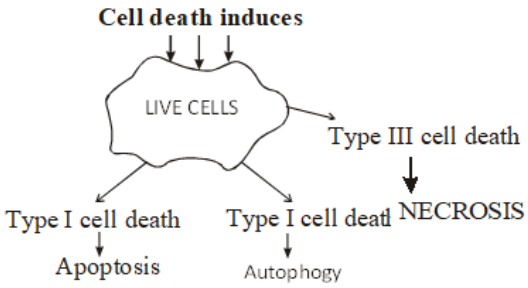
Pathways (Involved in Necrosis)



Treatment for Necrosis

1. Antibiotics
2. Antioxidant
3. Debridement surgical removal of deal tissues.
   If it is severe then amputation of limbs or organs.

Summary

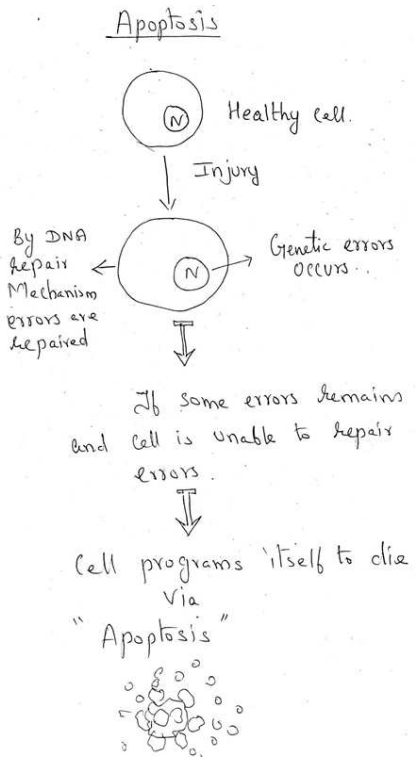


2.6 APOPTOSIS

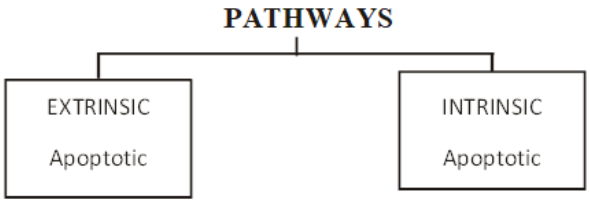
- Apoptosis is the process of programmed cell death.
- Biochemical events in the cell during Apoptosis
  a) Blebbing
  b) Cell Shrinkage
  c) Nuclear fragmentation
  d) DNA fragmentation
- **In humans (Adult):** 50-70 billion calls die due to Apoptosis in a day 8-14 yrs of age in children : 20-30 billion cells die in a day.

HISTORY

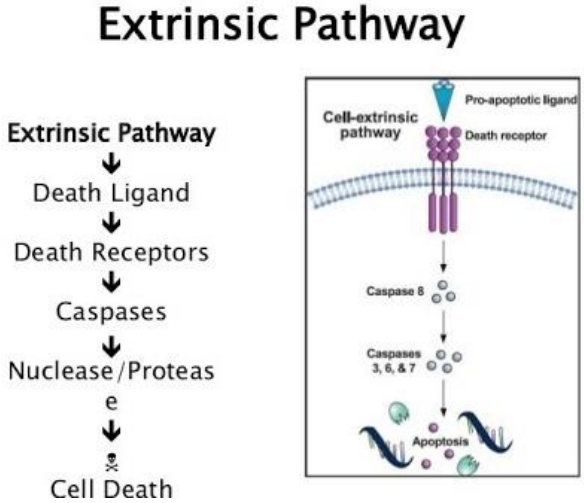
Carl vogt (1842) German scientist 1st described Apoptosis. The term "Apoptosis" 1st coined by Kerr (1972).



EXTRINSIC PATHWAY



- The cell in extrinsic signalling pathway produces Transmembrane death receptor which are members of Tumor necrosis factor (TNF) Super family.
- Death receptor binds to pro-apoptotic ligands such as Apo3L, Apo2L
- These interaction activities initiator caspases 8, enzyme which are proteares in nature & then activates caspases 3, 6 & 7.
- Lead to degradation of cellular function & finally causes Apoptosis.



INTRINSIC PATHWAY

The cell in Intrinsic pathway produces P53 protein, where NOXA produces PUMA & BAK converts into BAX.

⇩

BAX binds to Mitochondria

⇩

Produces now Cyt C + Apaf1 + Procaspase 9

⇩

Apoptosome formation

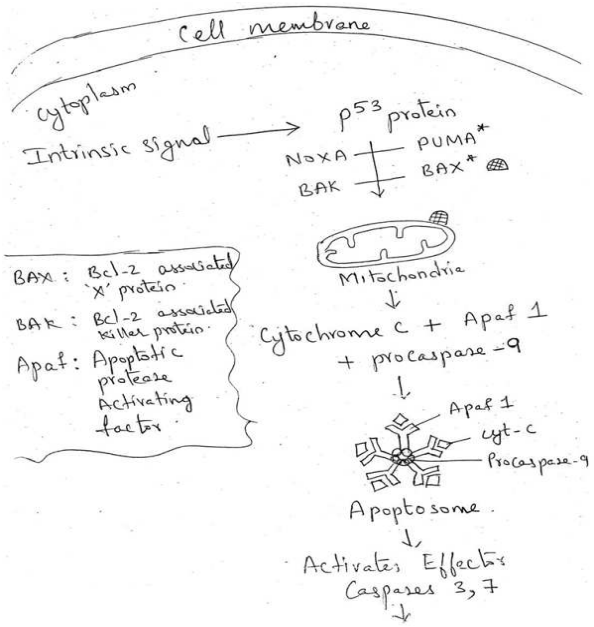
⇩

Activates Effector caspases 3,7

⇩

Leads to blebbing, nuclear fragmentation & Apoptosis

CELL MEMBRANE



UNIT III

PRINCIPLES AND MECHANISM OF INHERITANCE

3.1 MENDEL'S EXPERIMENTS

Gregor Johann Mendel is known as "Father of Genetics. He is the first person to discover the principles of genes, transmitting the traits from parents to off springs. In 1856, he began his Hybridization or breeding experiments with Garden pea (Pisum Satirum) plants.

Mendel selected Garden pea for Hybridization experiments for the following reasons.

1. Peas have many distinct, well defined and observable morphological charecteristics. (traits).
2. Pea plants are easy to cultivate. The plant size is small and many plants can be grown in small plots.
3. Peas are annual. Their complete life cycle from germination to flowering and to produce seeds with in one year.
4. Pea flowers are bisexual and naturally self fertilizing. Single plant has maternal and parent.
5. The flowers are large for easy emasculation (removal of stamen). Hence artificial cross fertilization is possible.
6. The off spring of cross fertilized plants are fully fertile.
7. A single pea plant produces many seeds to see all possible variations in the progeny. Mendel selected seven characters in garden pea plant. They are
   - **Flower position -** axial (or) terminal
   - **Pod colour -** green (or) yellow
   - **Pod shape -** Inflated (or) constricted
   - **Stem length -** Tall (or) short
   - **Seed shape -** Round (or) wrinkled
   - **Seed colour -** Yellow (or) Green
   - **Flower colour -** Purple (or) white.

Mendel Performed artificial cross fertilization between selected pairs. By removing male parts (stamen) before and dusting the stigma of emasculated flowers with pollen from selected plants.

Seven Chartcters in Pea Plant Studied By Mendel :

F_1 Crosses for Seven Charecters In Pea Plants				
Character	Dominant Trait	× Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower Color	 Purple	 White	705:224	3.15:1
Seed Color and Seed Shape	 Green	 Yellow	6,022:2,001	3.01:1
	 Round	 Wrinkled	5,474:1,850	2.96:1
Pod Color and Pod Shape	 Green	 Yellow	428:152	2.82:1
	 Inflated	 Constricted	882:299	2.95:1
Flower Position and Stem length	 Axial	 Terminal	651:207	3.14:1
	 Tall	 Dwarf	787:277	2.84:1

F1 crosses for seven characters in pea plants and their results in F2 generations

The results of Mendel's experiments are

1. The inheritance of each trait of an individual is determined by GENES.
2. For morphological charecteristics (traits), an individual inherits genes from each parent.
3. Males and Females contribute equally in passing the traits to the offspring.
4. A particular trait, may not appear in an individual, but still can pass on to next generation.

Mendel explained the factors for transmission of traits. These two principles are called MENDELS PRINCIPLES (OR) MENDEL'S LAW.

1. Law of seggregation - Mendel's 1st law
2. Law of Independant assortment - Mendel's 2nd law.

LAW OF SEGGREGATION:

For any perticular trait in an individual the pair of factors responsible for that trait, Separate during gametogenesis. And only one factor passes to an off spring.

LAW OF INDEPENDANT ASSORTMENT

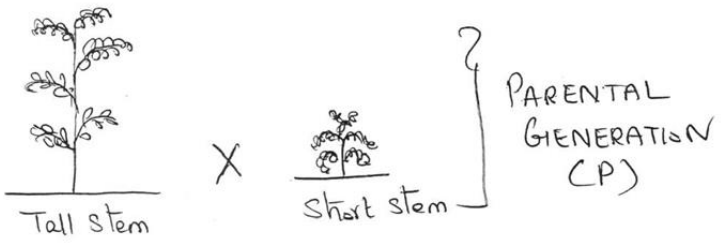
The inheritance of one trait doesnot affect the inheritance of another.

FACTORS CONTRIBUTING TO SUCCESS OF DMENDELS EXPERIMENTS

1. **SELECTION OF MODEL SYSTEM:** Plant pea is simple, several pure lines are available. Selection of well defined parents are available
2. **SELECTION OF TRAITS:** Mendel inherited only single trait during his experiments to get a clear idea.
3. **STUDY OF DISCRETE GENERATION:** Mendel carried out experiments for one generation only. But within generation he observed numerous plants.
4. **EXCELLENT RECORD KEEPING:** One of the strongest points in Mendel's experiments was Quantitative record keeping. Mendel noted down actual numbers of plants this made possible for statistical and mathematical analysis.

3.2 LAW OF SEGREGATION - MONOHYBRID CROSS / RATIO

1. A breeding experiment involving only one pair of contrasting charecters is called monohybrid cross.
2. Mendel selected the parents of two, having contrasting appearance for one charecter. For example: Tall stem and short stem.



Two different varieties of plants are considered as parental generation. (P). These two plants are artifially cross polinated to produce first filial generation also called as F_1 generation. The plants of F_1 generation wave sown in soil and allowed the next generation called second filial generation or F_2 generation.

1. Mendel crossed tall pea plants with short pea plants.
2. Monohybrid cross determined the inheritance of factors responsible for the height of the plant.
3. p generation comprises of tall charectors and short charector.
4. All plants in F1 generation are Tall.
5. It appeared that short charector was not present in plants.
6. Short charector reappeared in some plants in F2 generation.
7. Statistical analysis showed 75% tall and 25% short. in F2 generation

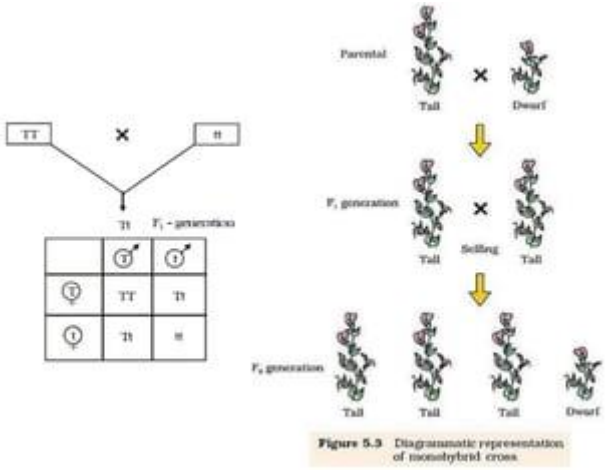


Figure 5.3 Diagrammatic representation of monohybrid cross

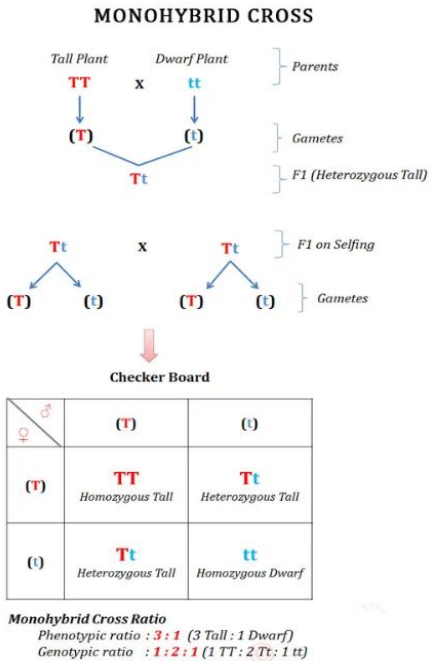
ANALYSIS OF MONOHYBRID CROSS

Mendel proposed that each charector was due to pair of factors.

Example: Tall (TT) : Short (tt)

The contrasting charecters for each trait are called Allelomorphs (or) Alleles.

MONOHYBRID CROSS



Allele expressed in Hybrid plant is termed as DOMINANT and alleles which do not express in hybrid is termed as recessive.

GENOTYPE : The composition of alleles for particular trait (Ex: TT, tt)
PHENOTYPE: Appearance of organism for particular trait. (Tall, short)

F1 generation has two dissimilar parents and received allele 'T' from Tall plant and allele 't' from short plant. Genotype of Hybrid plant is 'Tt'. and phenotype of hybrid plant is tall because T1 allele is dominant over allete 't'.

PUNNETT'S SQUARE OF MONOHYBRID CROSS - *Reginald Punnett*

		Maternal	
	Gametes ♀/♂	T	t
PATERNAL	T	TT (Tall)	Tt (Tall)
	t	Tt (Tall)	tt (Short)

PHENOTYPIC RATIO FOR MONOHYBRID CROSS — 3 : 1 — 3-Tall, 1-short
GENOTYPIC RATIO FOR MONOHYBRID CROSS — 1 : 2 : 1

LAW OF INDEPENDENTASSORTMENT

DIHYBRID AND TRIHYBRID CROSS inheritance of one trait doesnot affect the inheritance of another. Breeding experiments involving study of two pairs of contrasting charecters are called DIHYBRID CROSS. A cross involving three pairs of contrasting characters are called TRIHYBRID CROSS.

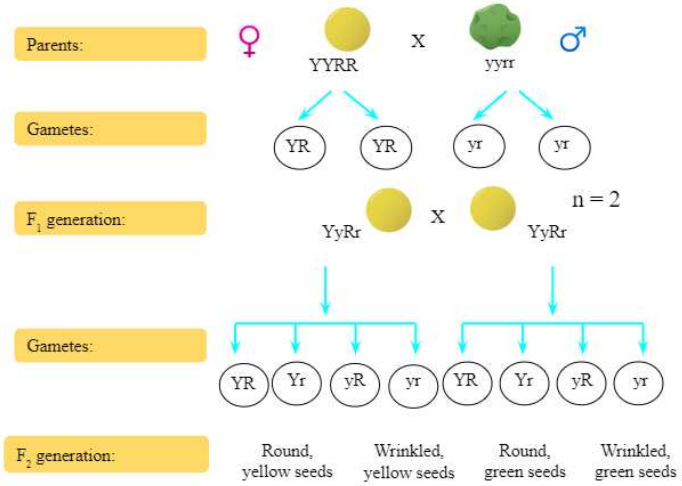
DIHYBRID CROSS

Mendel did an experiment where he crossed a pure - breeding plant bearing yellow and round seeds with pure - breeding plant bearing green and wrinkled seeds.

Yellow seeds are dominant over green seeds, and round seeds are dominant over wrinkled seeds. In F2 generation, four types of combinations were observed.

a) Both dominant trait in 9/16 of the population.
b) First trait dominant and second trait recessive in 3/16 of the population.
c) First trait recessive and second trait dominant in 3/16 of population.
d) Both traits recessive in 1/16 of the populates

DIHYBRID CROSS

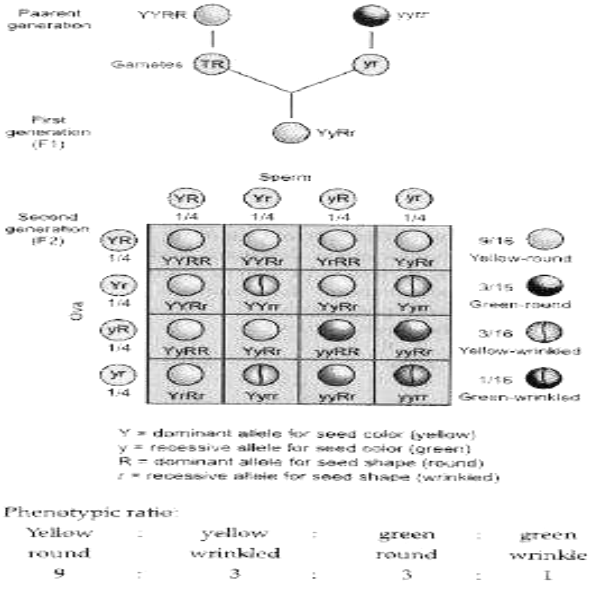


Type	Ratio	Combination
Yellow, Round	9/16	Parental
Yellow, Wrinkled	3/16	Recombinant
Green, Round	3/16	Recombinant
Green Wrinkled	1/16	Parental

DIHYBRID CROSS BETWEEN PLANTS WITH YELLOW ROUND SEEDS AND GREEN WRINKLED SEEDS

(P)	YYRR	X	yyrr
Gameter	YR		yr
F_1 Generation	YyRr		
F_1 plants are crossed	YyRr	X	YyRr
Gametes	YR	Yr	yR	yr

PUNNETT SQUARE FOR DIHYBRID CROSS



Phenotypic ratio:

Yellow round	:	yellow wrinkled	:	green round	:	green wrinkle
9	:	3	:	3	:	1

PHENOTYPIC RATIO : 9 : 3 : 3 : 1
Genotype Ratio : 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

1-YYRR, 2 - YYRr 1 - YYrr

2-YyRR 4 - YyRr 2 - Yyrr

1-yyRR 2 - yyRr 1 - yyrr

Mendel's law of Independant Assortment states. that, when dihybrid form gametas.

1. Each gametes receive one allele from each allelic pair.
2. The alleles are independantly assorted during gamete formation i.e. each allele of any one pair is free to combine with any allele from each of the remaining pairs during gamete formation.

Tri hybrid cross

A cross involving three pairs of contrasting charecters are called trihybrid cross Trihybrid cross between tall plants bearing yellow and round seeds and shokrt plants bearing green and wrinkled seeds.

Genotype for tall plants with yellow & round seeds - TTYYRR and

Genotype for short plants with green & wrinkle seeds - ttyyrr.

TRIHYBRID CROSS

[P] Parents TTYYRR X ttyyrr

Gametes TYR tyr

F1 Generation TtYyRr

Gametes : TYR TYr TyR TYr

tYR tYr tyR tyr

♀ / ♂	TYR	TYr	TyR	Tyr	tYR	tYr	tyR	tyr
TYR	TTYYRR	TTYYRr	TTYyRR	TtyyRR	TtyyRR	TtyyRr	TtYyRR	TtYyRr
TYr	TTYYRr	TTYYrr	TTYyRr	TTYyrr	TtyyRr	Ttyyrr	TtYyRr	TtYyrr
TyR	TTYyRR	TTYyRr	TTyyRR	TTyRr	TtYyRR	TtYyrr	TtyyRR	TtyyRr
Tyr	TTYyRr	TTYyrr	TTyyRr	TTyyrr	TtYyRr	ttyyRr	TtyyRr	TtyyrR
tYR	TtYYRR	TtYYRr	TtYyRR	TtYyRr	Ttyyrr	TtYyrr	ttYyRR	ttYyRr
tYr	TtYYRr	TtYYrr	TtyyRR	TtYyrr	ttyyRr	ttyyrr	ttYyRr	ttYyrr
tyR	TtYyRR	TtYyRr	TtyyRR	TtyyRr	ttYyRR	ttYyRr	ttyyRR	ttyyRr
Tyr	TtYyRr	TtYyrr	TtyyRr	ttyyRr	ttYyRr	ttYyrr	ttyyRr	ttyyrr

PHENOTYPE RATIO : 27 : 9 : 9 : 9 : 3 : 3 : 3 : 1

27: Dominant phenotype for 3 traits
Tall: Yellow: round

9: Dominent phenotype for 2 traits
Tall: yellow: wrinkled

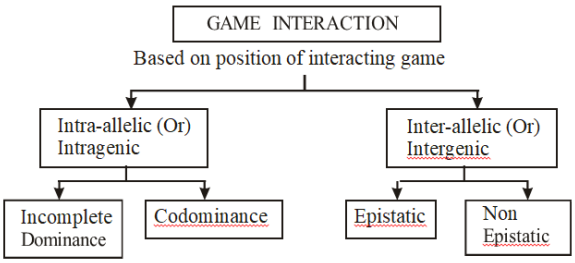
9: Dominant phenotype for 2 traits
Tall: green : round

9: Dominant phenotype for 2 traits
Short : yellow : round
3: Dominant phenotype for 1 trait
Tall: green : wrinkled
3: Dominant phenotype for 1 trait
Short : green : round
3: Dominant phenotype for 1 trait
Short : yellow : wrinkled
1: Dominant phenotype for 0 trait (or) Recessive phenotype
Short : green : wrinkled

3.3 Deviation from Mendel's Law:

Bateson and Punnett conducted several experiments to show that it is nokt necessary that single gene determines a singl,e charecter, but same charecter may be affected by more than one gene by Interaction. Such Mechanism of genes are call "GENE INTERACTION".

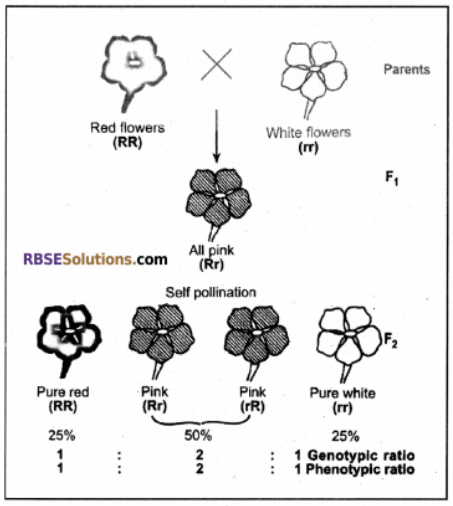
- **Intragenic :** Interaction of genes is between alleles of same locus.
- **Intergenic :** Interaction of genes is between alleles of different locus.



3.3.1 INCOMPLETE DOMINANCE

In this heterozygotes have intermediate phenotypes compared to dominant and recessive homozygotes. In such case instead of monohybrid classical ratio 3 : 1, modified

Monohybrid phenotypic ratio 1 : 2 : 1 are formed for example:

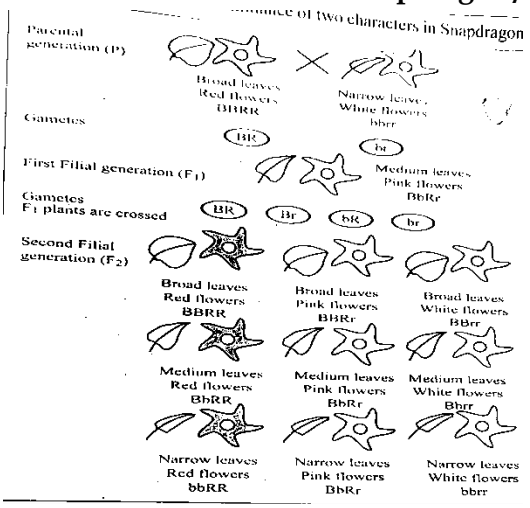


Incomplete dominance in *Mirabilis jalapa*

1. Incomplete dominance of one charecter of Mirabilus Jalapa

In F_2 generation, 25% plants gave red flowers, 25% gave white colour flower and 50% plants have pink flowers. Thus phenotypic ratio is modified frp, ,emde;s c;assoca; ratop 3"1 to 1"2"1. becaise mp allele is dominent over the other.

2. Incomplete dominance of two characters in snapdragon / Mirabilus Jalapa.



F_2 Generation - Punnett Square

	BR	Br	bR	br
BR	BBRR Broadd Red	BBRr Broad Pink	BbRR Medium Red	BbRr Medium Pink
Br	BBRr Broad Pink	BBrr Broad White	BbRr Medium Pink	Bbrr Medium White
bR	BbRR Medium Red	BbRr Meidum Pink	bbRR Narrow Red	bbRr Narrow Pink
br	BbRr Medium Pink	Bbrr Medium White	bbRr Narrow Pink	bbrr Narrow White

Modified Dihybrid Phenotypic Ratio:

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

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

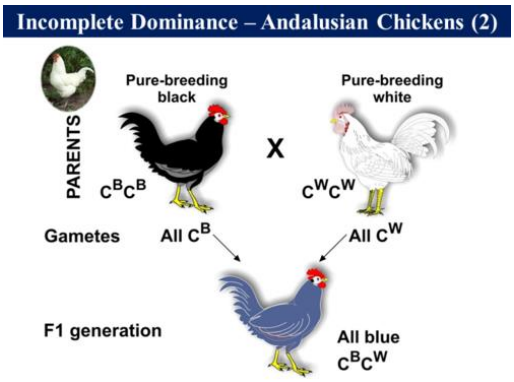
Incomplete dominance are called determining leaf shape and flower colour of snapdragen plants. A cross between plants with broad leaves and red colour flower and plants with narrow leaves and white colour flower gives plants with medium leaves and pink flowers in F1 generation.

Codominance

If the heterozygotes exhibits mixtone of phenotypic characters of both dominant and recessive characters of homozygotes, then the characters are said to be codominant and the phenomenon is called codominance.

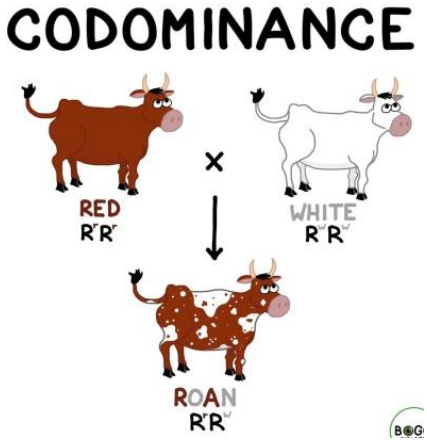
For Example:

1. Codominance in Andalusian Fowl



Homozygoes have either black or white but in F1 generation, the heterozygotes have blue feather, but in reality, the blue colour is a mosaic of black and white and appears to be blue.

2. Codominance in Cattle Coat Colour

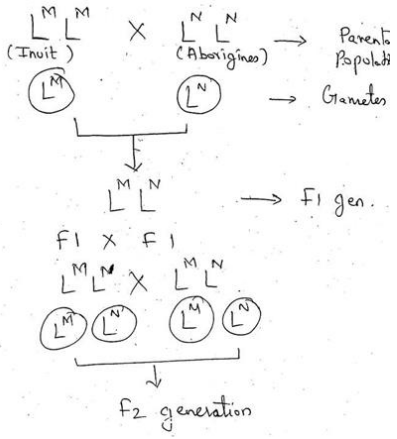


- 'Roan' coat showing red and white hair.
- In F1 generation, due to expression of both the alleles in heterozygoes, results in production of red and white hairs side by side forming roan coat colour.

3.3.2 CODOMINANCE

- Codominance is found in human genes. One best example in MN blood group. MN genes are under the control of Autosomal locus. MN genes expresses 'M' and 'N' ag. These are under the classification of Glycophorins.
- MN antigens binds on RBC surface.
- MN genes are operated by L gene located in chromosome 4. They contain 2 alleles.
- $L^M L^M$ & $L^N L^N$
- Codominance is best studied in 2 populations i.e.
- Invite population ($L^M L^M$)
- Aborigines ($L^N L^N$)

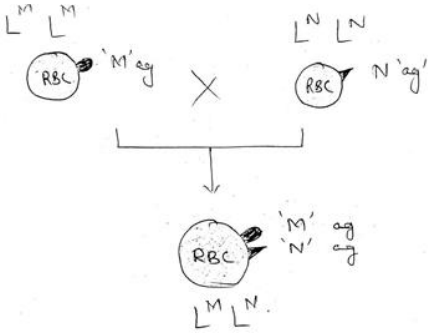
Crossing of Inuit & Aborigines :



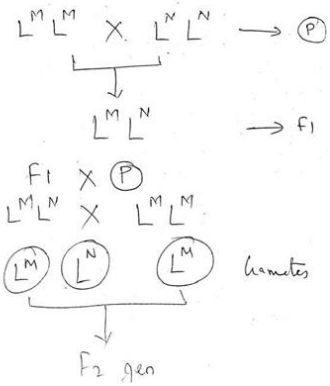
Phenotypic ratio 1: 2 :1

	L^M	L^N
L^M	L^ML^M Dominant Homozygotes RBC - 'M' Ag	L^ML^N Heterozygotes (RV - 'M' 'N'
L^N	L^ML^N Heterozygotes RBC $^{M}_{N}$	Dominant Homozygotes $(RBC)^N$

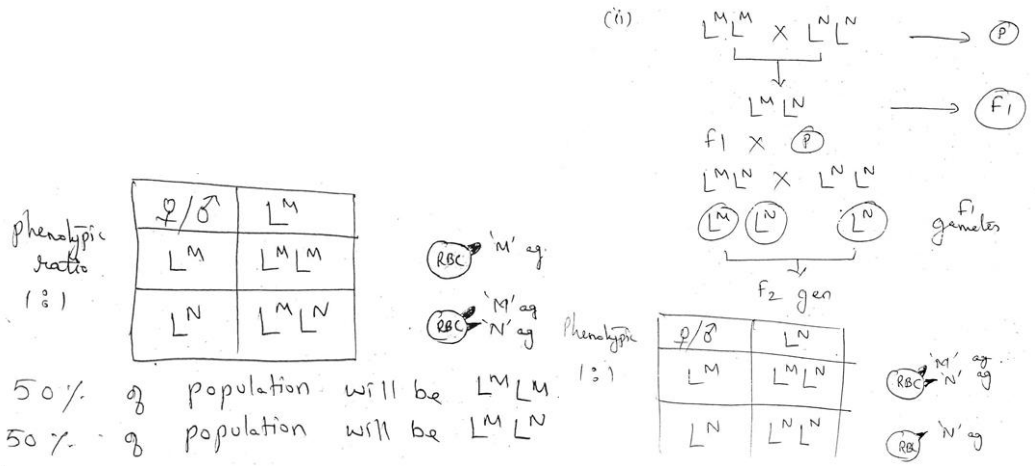
Punnett square for MN blood gp



TEST CROSS



Punett square for test cross



3.3.3 EPISTASIS GENE INTERACTION

Epistatic gene interaction describes how two independently assorted genes affect a single trait. This phenomenon are referred as EPISTASIS (or) Inter-allelic or Intergenic gene Interaction.

Mendels law of Independant Assortment states that alleles of two different genes are inherited independently resulting in charecteristic 9 : 3 : 3 : 1 ratio.
Modified dihybrid ratios are of following type they are,

1. DOMINANT EPISTASIS - 12 : 3 : 1
2. RECESSIVE EPISTASIS - 9 : 3 : 4
3. DUPLICATE DOMINANT EPISTASIS - 15 : 1
4. DUPLICATE RECESSIVE EPISTASIS - 9 : 7
5. DUPLICATE GENES WITH CUMULATIVE - 9 : 6 : 1
6. DOMINANT AND RECESSIVE EPISTASIS - 13 : 3

Epistasis in Greek means "Act of stopping". It is the phenomenon of gene interaction between Non-allelic genes, where one gene masks, suppresses or inhibits the expression other gene. The gene that is masking is called epistatic gene. The gene that is masked is called hypostatic gene.

1. DOMINANT EPISTASIS - (12 : 3 : 1)

It is also called Dominant suppressor interaction. {of the two genes in a dihybrid cross, let us consider, dominant allele are A and B, and recessive allele 'a' and 'b'). The gene which is masking are called epistatic and the gene which is masked are called Hypostatic. If A is epistatic over gene B, then dominant A masks not only 'a' but also 'B' and 'b'.

Example: Coat Colour In Dogs:

1. The two genes determining coat colour of dogs are 'I' and 'B'.
2. The Dominant allele 'I' inhibits pigment formation in for. The gene 'I' is epistatic. The recessive allele 'i' allows the expression of pigment in the for.

3. The gene 'B' is hypostatic, the dominant allele produces brown colour and the recessive (b) allele produces Black.

Dominant 'I' = inhibits pigment formation in the fur of dog.

Recessive 'i' = Allows pigment formation in the fur of dog.

Dominant 'B'= Produce Brown colour for in the dog.

Recessive 'b' = Produce Balck colour fur in the dog.

DOMINANT EPISTASIS SHOWN IN COAT COLOUR IN DOGS

Parental generation	White Dog	X	Brown Dog
Genotype	IIbb	X	iiBB
Gametes	(I) (b)		(i) (B)
F_1 generation	IiBb	(White)	
F_1 Gogs Crossed	IiBb	X IiBb	
Gametes	(IB) (Ib)	(iB) (ib)	
	(IB) (Ib)	(iB) (ib)	

Punnett Square

	IB	Ib	iB	ib
IB	IIBB DOMINANT MASKED	IIBb DOMINANT MASKED	IiBB DOMINANT MASKED	IiBb DOMINANT MASKED
Ib	IIBb DOMINANT MASKED	IIbb DOMINANT MASKED	Ii Bb DOMINANT MASKED	Ii bb DOMINANT MASKED
iB	Ii BB DOMINANT MASKED	Ii Bb DOMINANT MASKED	i i BB RECESSIVE DOMINANT	i i B b RECESSIVE DOMINANT
ib	Ii Bb DOMINANT MASKED	Ii bb DOMINANT MASKED	ii Bb RECESSIVE DOMINANT	i i b b RECESSIVE RECESSIVE

DOMINANT MASKED : 12 (White colour fur)

RECESSIVE DOMINANT : 3 (Brown colour fur)

RECESIVE RECESSIVE : 1 (Black colour fur)

PHENOTYPIC RATIO FOR DOMINANT EPISTASIS IS 12 : 3 : 1

The Dominant 'I' masks 'i', 'B' and 'b'. Hence the dihybrid ratio are modified from classical dihybrid ratio.

Parent genotypes	IIbb	X	iiBB
Phenotype	White		Brown
Gametes	Ib		iB
F1genotype		IiBb	
Phenotype		White	
Dihybrid cross	IiBb	X	IiBb

F2 genotypes	I-B-	I-bb	iiB-	Iibb
Phenotype	White	White	Brown	Black
Ratio	9/16+3/16=12/16		3/16	1/16

2. RECESSIVE EPISTASIS: (9 : 3 : 4)

In this mode of epistasis, one gene is epistatic over the other gene, but recessive allele is epistatic and is inhibitory to the expression of hypostatic gene.

Example: Coat colour in Mice.

1. Coat colour is determined by two genes
   a) Agouti - Agouti gene A
   b) Black - Black gene C
2. Three different colours are formed by the combination of there two genes.
3. Agouti colour is due to agouti gene A, which controls the distribution of yellow & black pigments on the hairs of mice and thus gives the hairs a charecteristic banded pattern. Hairs are black with yellow bands hear the tip of the hair.
4. Black colour is due to black gene C If Agouti gene is recessive, i.e. 'a' with combination of dominant C, then the hairs are solid black.
5. Albino colour is due to homozygous recessive for 'cc'. All mice with 'cc' genotype will be albino, even if they have dominant allele for 'A'. Albino mice lacks pigmentation and thus be white in colour.

Parent genotype	AACC	X	aacc
Phenotype	Agouti		Albino
Gametes	AC		ac
F1 genotype		AaCc	
Phenotype		Agouti	
Dihybrid cross	AaCc	X	AaCc

F2 genotypes	A-C-	A-cc	aaC-	aacc
Phenotype	Agouti	Albino	Black	Albino
Reason	C makes piment, A makes banding	cc is epistatic over A	C makes pigment, nc banding	cc is epistatic over A
Final phenotype	Black	Agouti	Albino	
	9/16	3/16	3/16+1/16=4/16	

1. Agouti (A-C) - 9 (Dominant Dominant)
2. Black (aa-C) - 3 (Recessive masked)
3. Albino (A-cc) - 3 (Dominant masked (or) Recessive)
   (aa cc) - 1 (Receswsive masked)

∴For recessive spistasis, the ratio is 9 : 3 : 4.

The recessive 'aa' are masked by 'Cc' and 'cc'.

b. DUPLICATE DOMINATE EPISTASIS

This made of epistasis are also called "duplicate action". The presence of dominant allela, either at a laws or at 'B' locus produces the same phenotype. Presence of either one of the dominant allele produces same phenotype.

Example: Seed shape in Sheperd's purse plant

1. There are two types of seed shape in shaperd's purse plant.
   a) Triangular
   b) Elongated or top shaped (▼)
2. The genes controlling these charectors are present at two different loci in two different chromosomes. The genes responsible for triangle shape of seed are expressed by either Dominant allele 'A' or dominant allele 'B'. Absence of dominant allele (or) Presence of recessive homozygous allele produces alongeted or top seed shape (aa, bb).

Parent genotype	AACC	X		aacc
Parent genotype	TTDD	X		ttdd
Phenotype	Triangular			Elongated
Gametes	TD			td
F1 genotype		TtDd		
Phenotype		Triangular		
Dihybrid Cross	TtDd	X		TtDd
F2 genotype	T-D-	T-dd	ttD-	Ttdd
Phenotype	Triangular	Triangular	Triangular	Elongated
Final Phenotype	9/16+3/16+3/16=15/16			1/16

	AB	Ab	aB	ab
AB	AABB (Triangle)Δ	AABb (Triangle) Δ	AaBB (Triangle) Δ	AaBb (Triangle)Δ
Ab	AABb (Triangle) Δ	AAbb (Triangle) Δ	AaBb (Triangle)Δ	Aabb (Triangle)Δ
aB	AaBB (Triangle)Δ	AaBb (Triangle)Δ	aaBB (Triangle)Δ	aaBb (Triangle)Δ
ab	AaBb (Triangle) Δ	Aabb (Triangle)Δ	aaBb (Triangle)Δ	aabb (Elongated)

A - B
A - B } the phenotype are 15 expressed
A - b

A - b } the phenotype are not expressed 1

Hence the duplicate dominant epistasis modified dihybrid ratio are 15 : 1.

Duplicate Recessive Epistasis: 9 : 7

This is also known as complementtion interaction. The two genes interact to produce a single phenotype. The phenotype are expressed in the presence of both the dominant allele. That is, the final phenotype are produced by the action of two independant gene. Thus modified dihybrid ratio are formed.

Example: Flower Colour of Sweet Pea : Dominant colour of sweet pea flower are purple. This colour is dependant on two non-allelic complimentary genes. They are

1) Gene C 2) Gene P.

The product of gene 'C' is an colour less substance. The product of gene 'P' transforms colourless substance to purple colour. This means both gene 'C' and gene 'P' are required for purple colour formation.

Parent genotype	CCpp	X		ccPP
Phenotype	White			White
Gametes	Cp			cP
F1 genotype		CcPp		
Phenotype		Purple		
Dihybrid Cross	CcPp	X		CcPp
F2 genotype	C-P-	C-pp	ccP-	CcPp
Phenotype	Purple	White	White	White
Final Phenotype	9/16	3/16+3/16+1/16=7/16		

♀/♂	CP	Cp	cP	cp
CP	CCPP (Purple)	CCPp (Purple)	CcPP (Purple)	CcPp (Purple)
Cp	CCPp (Purple)	CCpp (White)	CcPp (purple)	Ccpp (White)
cP	CcPP (Purple)	CcPp (purple)	ccPP (White)	ccPp (White)
cp	CcPp (purple)	Ccpp (white)	ccPp (white)	ccpp (white)

Thus for duplicate recessive epistasis the modified dihybrid ratio is 9 : 7.
Where 9 shows purple flower and 7 shows white flower in sweet pea plant.

13 - heterozygous chickens are white plum

3 - heterozygous chickens are coloured (21 plumage.

Inhibition of colour formation (due to presence of dominant I) and absence of colour (due to absence of dominant allele at 'C'), both produces white plumage as phenotype. To produce coloured plumage, the genotype at first locus should be ii and second locus should be 'C'.

Duplicate Gene with Cumulative or Effect: 9 : 6:1

The two genes are interacted to high both dominant allele produce same amount of contribution to the phenotype

Example: Coat colour swine

AA→Produces sandy coat colour

BB→ Produces sandy coat colour

AB→ Produces Brown coat colour

ab→ Produces No colour i.e White

Parent genotype	AABB	X		Aadd
Phenotype	Red			White
Gametes	AB			Ab
F1 genotype		AaBb		
Phenotype		Red		
Dihybrid Cross	AaBb	X		AaBb
F2 genotype	A-B-	A-bb	aaB-	aabb
Phenotype	Red	Sandy	Sandy	White
Final Phenotype	9/16	3/16+3/16=7/16		1/16

3.4 PENETRANCE & EXPRESSIVITY

The expression of an allele depends on two important factors. They are.

1) PENETRANCE

2) EXPRESSIVITY

PENETRANCE

The ability of an allele to express in an individual is called penetrance. Percentage of an individual exhibiting phenotype associated with Genotype, are always referred as penetrance. Penetrance of many alleles are variable, because phenotype is always influenzed with various factors such as

- Environmental factor
- Suppressor gene
- Epistatic gene etc.

Penetrance based on variations in phenotype for a genotype are classified into two.

1) COMPLETE PENETRANCE
2) INCOMPLETE PENETRANCE

Uniform Expressivity:

If gene expresses itself uniformly in all the individuals, it is said to have uniform expressivity. All the factors selected by Mendel for his experiments like

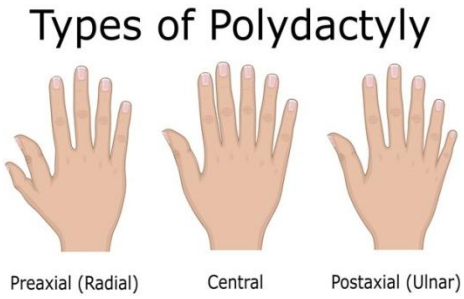
green or yellow pod, violet or white flower had uniform expressivity. For example, if one compares violet colour flower from number of plants, all flowers have same shade of violet. There will not be deep violet or pale violet flowers.

Variable Expressivity:

- Variation in expressivity is due to the presence of complementary, supplimentary or modifier (or) suppressor gene.
- Example: The ability of human beings to taste bitter substance phenyl this carbamide or
- C. some people detect the bitter taste at 0.16 mg/l of PTC, where as same at 130 gm/l of PTC.
- The gene for polydactyly, results in production of more than 5 fingers or toes per hand or foot & has 70% penetrance.

Human Polydactly:
- -Postaxial (little finger)
- -Preaxial (Thumb)
- -Central

In this case, extra thumb or extra little finger were observed.



Variation in penetrance occurs due to gender, age, environment, nutrient level, lifestyle, presence of interacting gene etc.

In Greek

Poly: Many

Dactyles: Digits →Complete toe / finger
→Partially formed
→Small mass (nubbin) okf soft tissue

Syndromes associated:

- Down syndrome, syndactly (joint fingers) etc.

WARDENBERG SYNDROME

- Best example of variable expressivity
- It is rare genetic conditions
- It is characterised as following

Type 1 : (WS-1) posses

- Cngenital hearing loss
- Forelock of white hair. (poliosis)

- Different colored eye (both eyes)
- Hetero chromic iridum
- Different color eye (multiple eye)
- Sectoral heterochromia iridum
- Patches in skin depigmentaiton
- Wider gap between innes carners of eyes - Telecanthus.
- High Nasal bridge.
- Flat nose tip
- Smaller edges of the noshils.

Type-2 (WS-2)

- Smaller eyes (microphthalmia)
- Hardeneal bones (osteopetrosis)
- deafness.
- Under development of inner - ear structure.
- A nosmia (lack of sense of small) due to back of olfactory bulb in the brain.

Type - 3 (WS-3) (or) Klein - Weardenburg Syndrome.

Same like type 1 with additional symptoms like affects in arms & hands.

- Camptodactyly - fingers bent
- Syndactyly - 2 fingers join
- Microcephaly

Type - 4 (WS-4) Shah - Wear henbari, syndrome)

Same type 2 with additional symptoms like lack of Nerves in intestines leading to bowel dysfunction and rarely claft lip.

PLEIOTROPISM

A classical Mendelian trait is that one gene in genotype is expressed as a single character in the phenotype. But this is not for many genes, because.

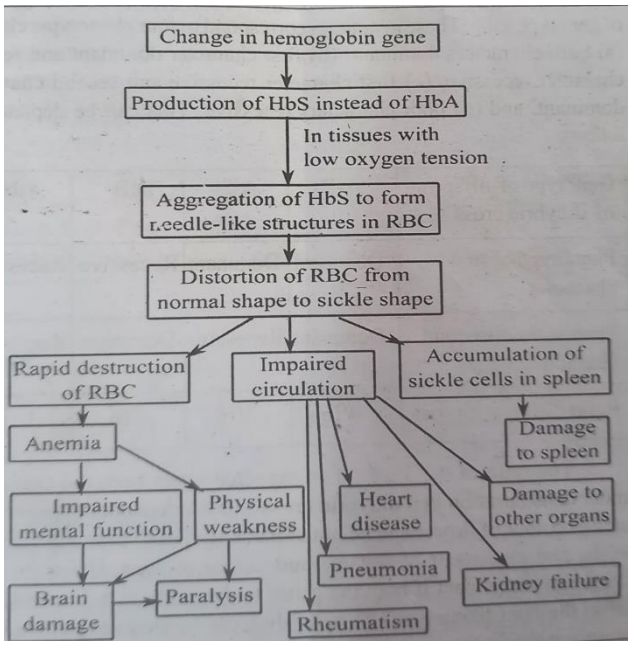
- A single gene may affect more than one trait.
- A single trait may be produced by more than one gene.
- The expression of trait is affected by environmental factors.

The phenomenon of single gene influencing more than one charector is called pleiotropic and such gener are called pleiotropic genes.

Examples for pleiotropism are

1. Sickle cell anemia
2. Phenyiketoneuria
3. Recessive genes for vestigial wings in drosophila.

Sickle cell anemia is a genetic disease results in premature death. The disease is due to defect in gene that produces Hemoglobin. This defective Haemoglobulin forms needle like crystals in RBC, which makes the cell discoid shape. The deformed RBC are destroyed leading to Anemia.



PHENOCOPY

- The term coined by Richard Gold sehmidt in 1935.
- Phenocopy is a variation in phenotype which is caused by environmental conditions.

EXAMPLES

- Microcephaly
- It is rare neurological disorder or condition in which infant's head is significantly smaller than heads of other children of same age and sex.
- It is usually congenital
- Causes :
  1) **Craniosynostosis:** Premature fusing of the joints between bony plates
  2) Chromosomal abnormalities: Down syndrome
  3) Decreased oxygen to the fetal brain (Cerebral anoxia)
  4) Infections passed to fetus during pregnancy.
  5) Malbition
  6) Exposure to drugs, alcohol.

Signs & Symptoms

- Head circumferance is smallest than normal. Dwarfism.
- Short stature Facial distortions
- Balance & coordination problem Hyper activity
- Neurological problem Nika viral Infection
- Treetment
- Amino acid replacement therapy

CLEFT LIP

1) Cleft lip (or) cleft palate are openings or splits in upper lip, the roof of mouth (palate) or both.
2) They are common birth defects.

Symptoms

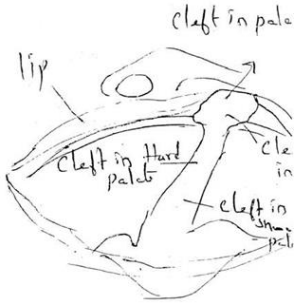
- Difficulty with feeding
- Difficulty swallowing
- Nasal speaking voice
- Chronic ear infertions

Risk Factors

1) Family history
2) Exposure to certain substances during ncy.
3) Having Diabetes
4) Being abese during Pregnancy

Complications:

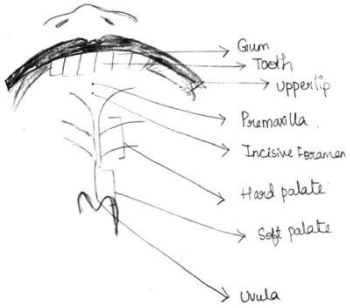
- Difficultly in feeding
- Ear infections
- Hearing loss
- Dental problems
- Speech difficulties



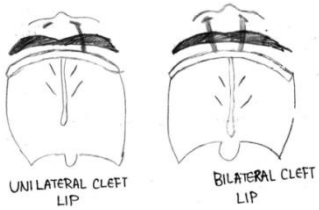
Preventions :

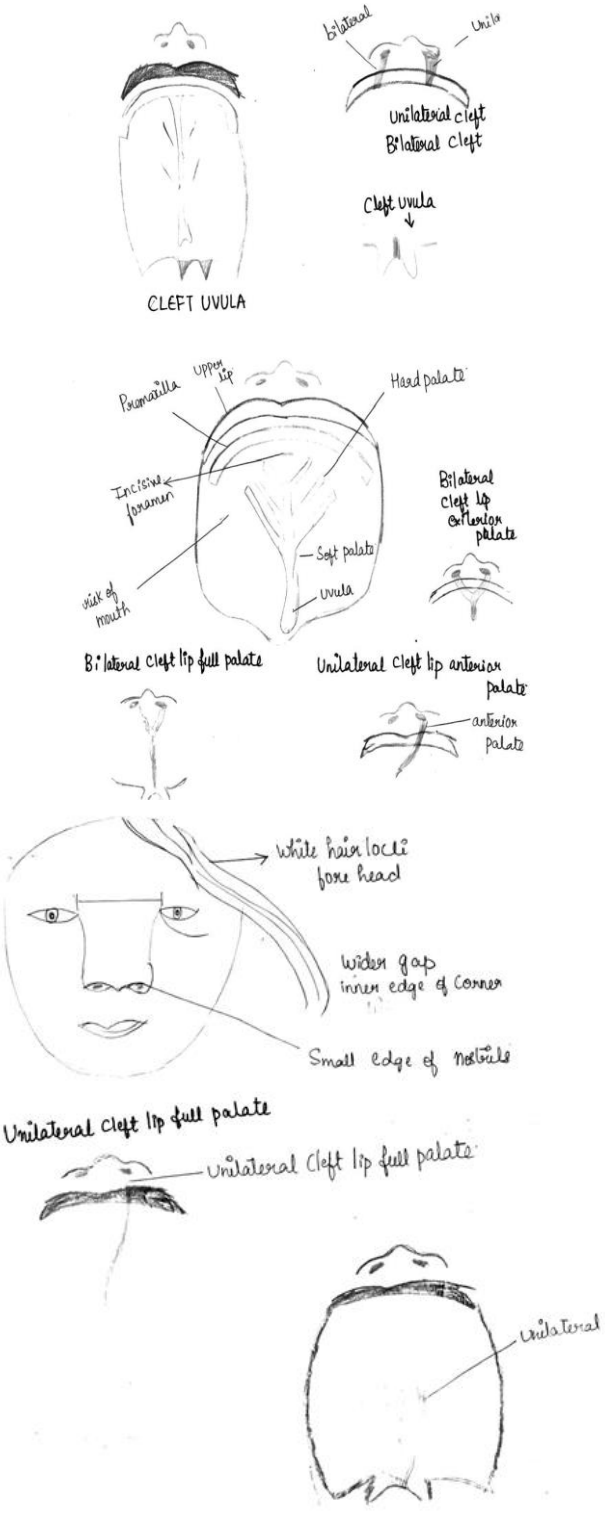
- Considers genetic counseling
- Take prenatal vitamins
- Do not use tobacco (or) alchol

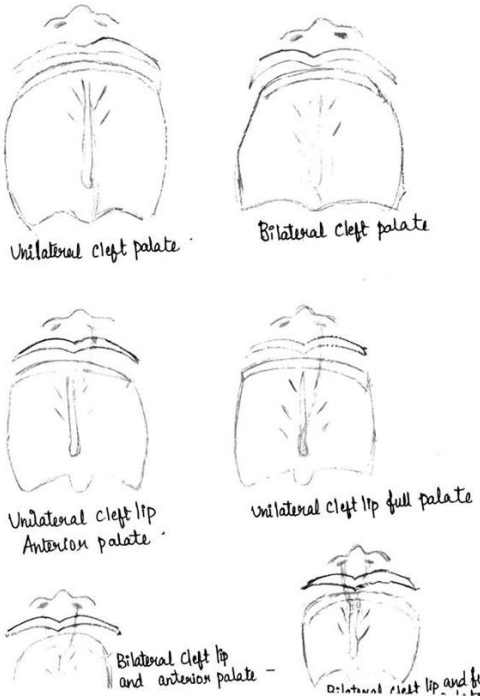
CLEFT LIP



Types of Cleft Lips




Bilateral
Uvula
Unilateral cleft
Bilateral Cleft
Cleft Uvula
CLEFT UVULA
Premaxilla
Upper lip
Hard palate
Incisive foramen
Soft palate
Uvula
Bilateral Cleft lip & anterior palate
Bilateral Cleft lip full palate
Unilateral Cleft lip anterior palate
anterior Palate
White hair locli fore head
Wider gap inner edge of corner
Small edge of nostrils
Unilateral Cleft lip full palate
Unilateral Cleft lip full palate
Unilateral



3.5 MULTIPLE ALLELEISM

An allele of a genetic locus having more than two allelic forms with ina population i.e more than one allele code for a gene. If more than 2 allele governing the same character, then it is called Multiple alleles.

Sample:

1. Multiple alleles in Rabbits for loat color 4 alleles exist in 'C' gene.
   wild - type version - C+ C+ expressed as Brown - for chinchilla phenotype - C^{ch} C^{ch} expressed as Black tipped white fur
   Himalayan phenotype C^h C^h Black fue on extremities & white for Albino, recessive phenotype - cc expressed with white fur.
   Albino can also be expressed as C^a C^a .
   Coat colour of Rabbit: It is inherited as a series of multiple alleles. In case of coat color of Rabbits, there are 4 aleles and each one is expressed with a different phenotype.

The order of Dominance is C> C^{ch}>C^h>c^a

Example: Himalayan coat color Rabbit crossed with same genotype (hetarozygotes)

C^h C^a X C^hC^a------(p)

(C^h)(C^a) (C^h)(C^a)------- Gemetes

1st Cross

C^hC^a→ F_1

F_1 X F_1

C^h C^a C^h C^a

	C^h	C^a
C^h	C^hC^b (H)	C^hC^a (H)
C^a	C^hC^a (H)	$C^aC^{a(A)}$

H-Himalayan
A-Albino

Eye color of Drosophila

Red eyed Female X Red eyed Female

WW X ww

X X X

X

1st cross

F_1

	W	W
w	Ww0	Ww0
w	Ww	Ww

→Red Eye Female
→Red

Red eyed female X red eyed male

Ww X ww

X X x

♀

		X 0 W	X 0 w
X	W	WW Red eyeFemale	WW Red eyeFemale
Y	w	Ww Red eyeMake	ww White eyeMale

♂

- Multiple alleles in eye color of Drosophila and 14 alleles for eye color.
- Ominant eye color is Red cessive eye color is white
- But Heterozygates produces other shades from white to red i.ewine, blood, eosin, honey, pearl,lrory.
- In F2 generation, Mutant strains show variation in wide range color between red to white.

ABO Blood Group:

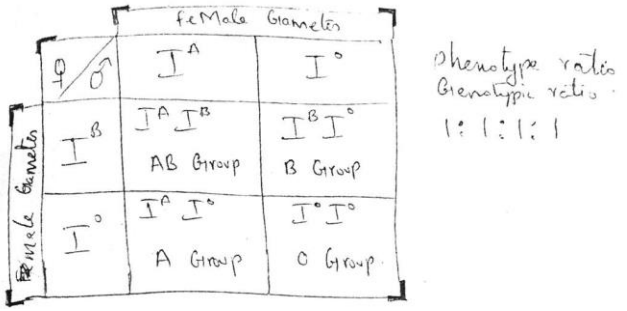
Blood groups are inherited from both parent. There are three alleles for ABO blood groups in humans. If chromosome contains allele A, then protain A is prokduced, then the RBC's of that individual will contain protein A on its surface. And that individual falls under 'A' Blood group like wise for allele B, AB. In 'O' blood group, the RBC's carry no protein on its surface.

Types of blood groups & its Genotype

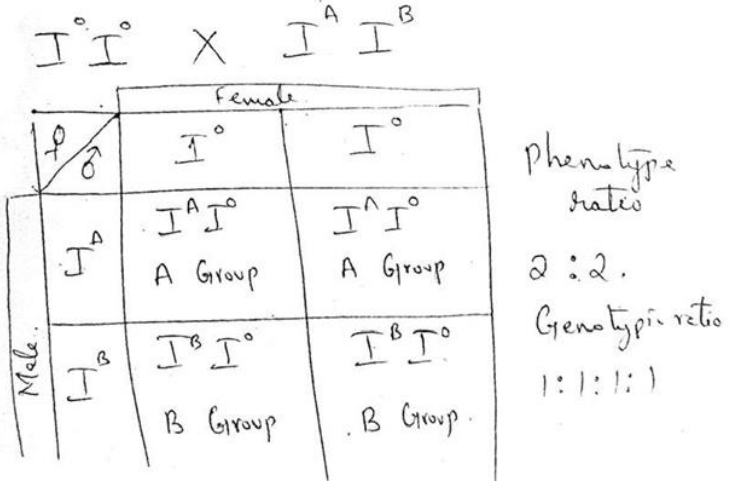
A	$I^A\ I^A$ $I^A\ I^o$	RBC
B	$I^B\ I^B$ $I^B\ I^0$	RBC
AB	$I^A\ I^B$	RBC 'A' 'B'
O	$I^0\ I^0$	RBC

Example: When heterozygotes of group A( ) crosses with heterozygotes of group B( ). Find out the genotype in 1st cross.

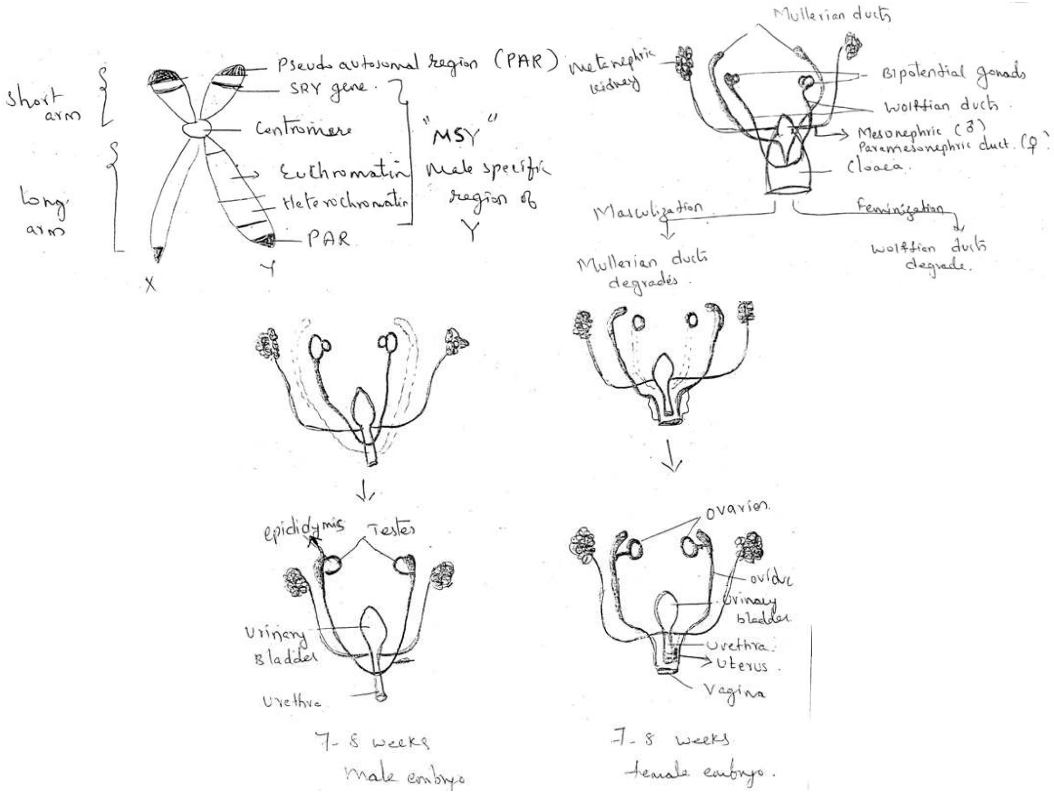
$I_A\ I_0$ X $I_B\ B_0$-------- (P)



Example: When Homozygote of O group (♀) cross with group AB. Find out the possible blood groups in the off spring.



3.6 XY Chromosome



Sex-determining region of Y chromosome in embrgonic germ cells (SRY gene)

⇩

SRY protein - i.e. Testis determining protein

⇩

This causes gonad medulla to differentiate into testis

⇩

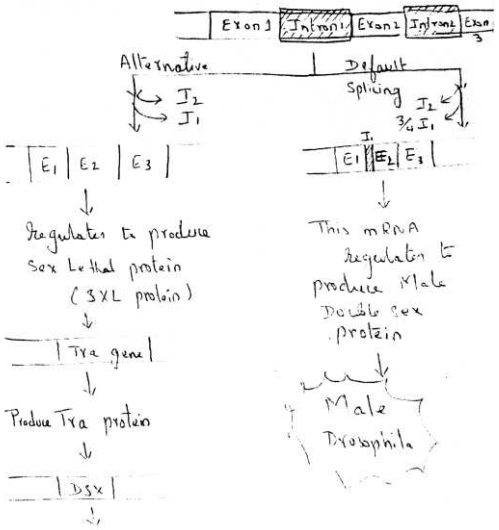
Leycling cells	Sertolicells
⇩	⇩
Secretes	Secretes
⇩	⇩
Testosterone Dihydro testostenone	Anti mullerian hormone
⇩	⇩
Develops epididymis seminal vesicle vas deferens	Develops fallpian tube, utenis vagina

DHT➔ develops porstate glands & penis

3.6.1 Alternative Splicing

When a single 10 transcript (mRNA) is processed into two or more different ways then it is called as Alternative splicing sex determination in Drosophila is the

best example in Alternative splicing. In this splicing to produce male drosophila, some part of intron 1 retains in mRNA. This mRNA produces male double sex protain. While in producing female drosophila, complete removal of I_1 & I_2 makes the mRNA to produce SXL protein. This protain activates 'Tragen & produce traprotain. Lator Tra protain regulator DSX gene to produce female double sex proteins . This mRNA regulation to produce femate double sex protein.



3.6.2 Sex Determination in Human

- Sexual differentiation in humans is the process of development of sex differeness in humans.
- Sexual differentiation includes development of different genitalia and the internal genital tracts and body hair play a role in gender identification.

XY Sex Determination System

This system is responsible for the development of phenotypic differences between male & female human from undifferentiated zygote. i.e.

Female	Contains	XX	Chromosome
Male	Contains	XY	Chromosome

During early embryonic development, both sexes possess equivalent internal structures. i.e

Femalie - para Mesonephric ducts

Male - Mesonephric duets

Until seven weeks after fertilization, the fetus appeors to be sexually indifferent, i.e. the fetus neither looks like make or female, but over next 5 weeks, the fetus begins to produce Harmones thatcauses sex orgens. This process is called sexual differentiation.

In males the Y chromosome carry essential gene called SRY gene. (Sex - determining region Y) which produces SRY protein.

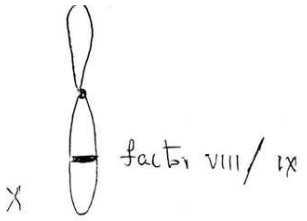
3.6.3 X- Linked Inheritance

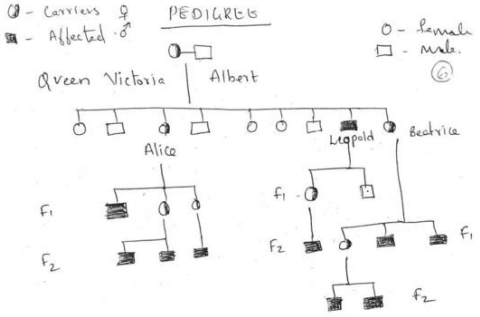
HEMOPHILIA:

It is bleeding disorder, which slow blood clotting process. i.e., Prolonged bleeding or oozing after injury, surgery or sometimes spontaneous bleeding 2 types of hemophilia.

1. **Hemophilia A** (Classic hemophilia or clotting factor gene factor VIII deficiency) is inactivated
2. **Hemophilia B** (Christmas disease or factor IX deficiency) (clotting factor IX gene is inactivated

Example: The "royal disease", a blood disorder transmitted from queen victoria to European royal families is striking example of X-linked recessive inheritance.



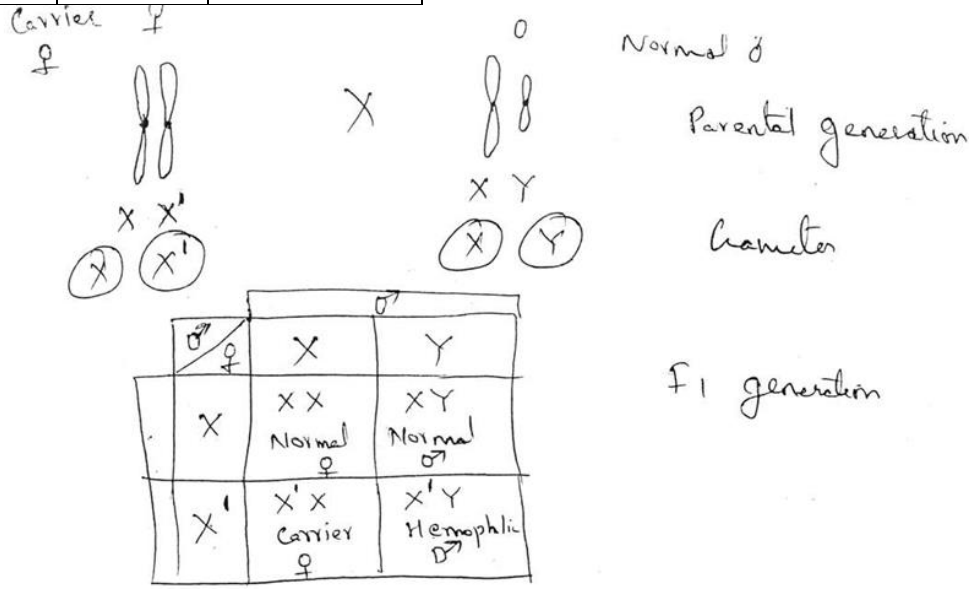


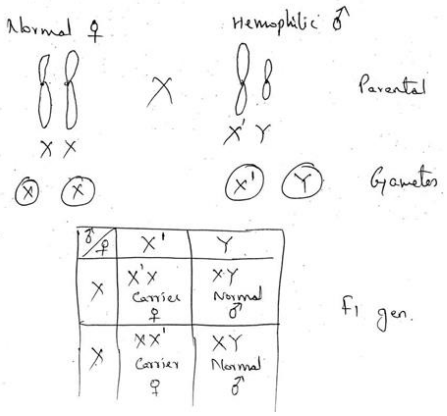
When assigning alleles for sex linked traits for haemophilia.

X^H - Dominant (X)

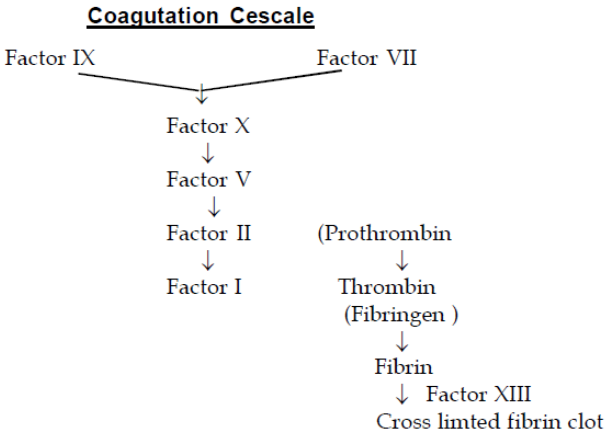
X^h - recessive (X^1)

	Normal	Hemophitia
Female	$X^H X^H$ $X^H X^h$	$X^h X^h$
Male	$X^H Y$	$X^h Y$

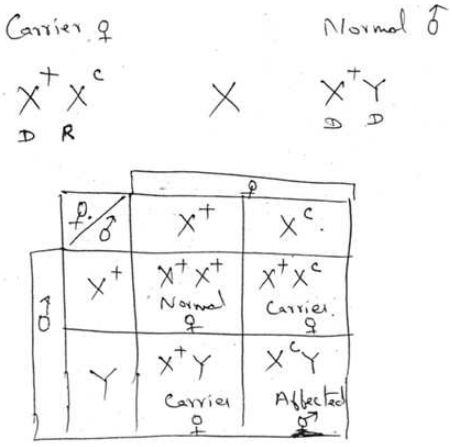




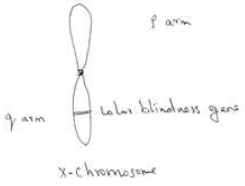
Coagutation Cescale



3.6.4 Color Blindness (Red - Green Colour blind)



Color blindness is X - linked recessive inheritance. Red - Green color blindness mean that a person cannot distinguish shades of red and green, but their ability to see is norma.

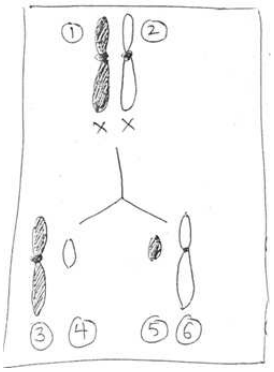


Males are affected more than females.

- X^{+} or X^{N} is dominant
- **X^{c} or X^{n} is recessive**

3.6.5 X - Inactivation or XCI (X- Chromosome inactivation)

It is also called Lyonization after English Geneticist Mary Lyon it is the process by which one of copies of X chromosome is inactivated therian female mammals. (Therian means dominant mammals which gives birth to live young ones without a shelled egg).

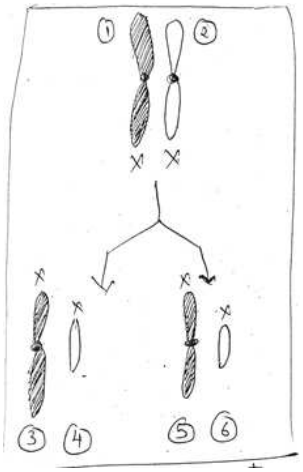


1. Maternal X chromosome
2. Paternal X chromosome
3. Maternal X chromosome is normal while
4. Paternal X chromosome is inactivated
5. Inactivated Maternal X chromemosome
6. Normal pateral X chromsome

In both the offsprings, paternal X chromosome inactivated i.e. (4) & (6)

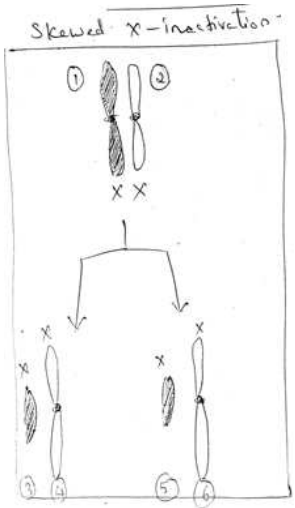
1. Maternal X chromosome

2. Paternal X chromosome



3. & (6) Normal maternal X chromosome (3) & (5), maternal X - chromsome inactivate

Paternal X - chromome is normal (1), (4), (6)



X-inactivation Example For Random - Calico Cat

Heterozygovs : Black & Orange

CAT (X^B) (X^0)

$X^0 X^B$

$X^b X^b$ ➔ Black females cat

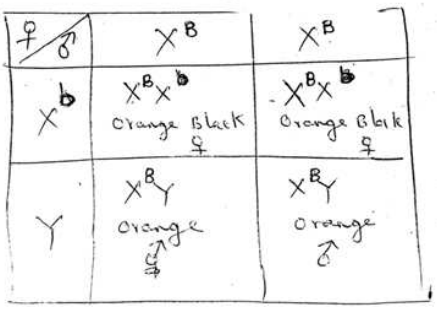
$X^B X^B$ ➔ Orange females : cat

$X^B Y$ ➔ Orange males cat

$X^b Y$ ➔ Black male cat

$X^B X^b$ ➔ Black & orange cat female (calico cat)

$X^B X^B$ (orange cat) — $X^b Y$ (Black cat)



Mechanism

X-inactivation involves transcriptional silencing of one of the two X-chromosome in female mammals.

⇩

This process is controlled by

X - inactivation centers (X ic) at q arm , gene X IST controls.

X- inactivation (X inactive specific transcript)

⇩

∴Ultimate goal of X inactivation is

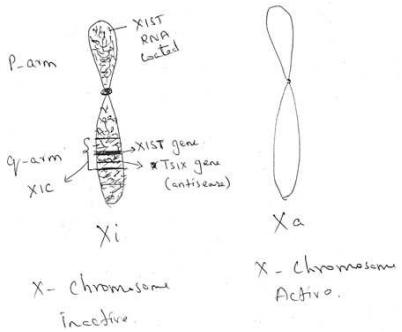
Dosage compensation i.e. describes the process in which organisms equalize the expression of genes between members of different sexes.

⇩

∴Females are mosaic for expression of X chromosome linked genes.

Types of X - inactivation

1. Raddom X - inactivation ➔ either X or X
2. Imprinted X - inactivation ➔ skewed particularly



XIST gene produces RNA which binds to X - chromosome & inactivates.
TSI X gene produces antisense RNA to inactivate XIST.

UNIT IV

LINKAGE, RECOMBINATION & EXTENSION TO MENDEL'S LAWS

4.1 LINKAGE AND RECOMBINATION

Discovery of Linkage

T.H.Morgan in 1910, while working with drosophila proposed that couppling and repulsion are the aspects of same phenomenon called as LINKAGE.

COUPLING AND REPULSION THEORY

1. **COUPLING:** It is defined as tendency of allelen either dominant or Recessive coming from same parent enter the same gametes in highes proportion and get inherited together.
2. **REPULSION:** It is defined as tendency of alleles either dominant or recessive coming from different parents to repel and enter different gametes and get inherited separetely.

The concept of linkage, proposed by Morga, replaces coupling and repulsion hypothesis the salient features of concept of linkage are

a) Genes are arranged in linear pattern on the chromosomes.

b) The number of genes on chromosome are more than the number of chromosomes.

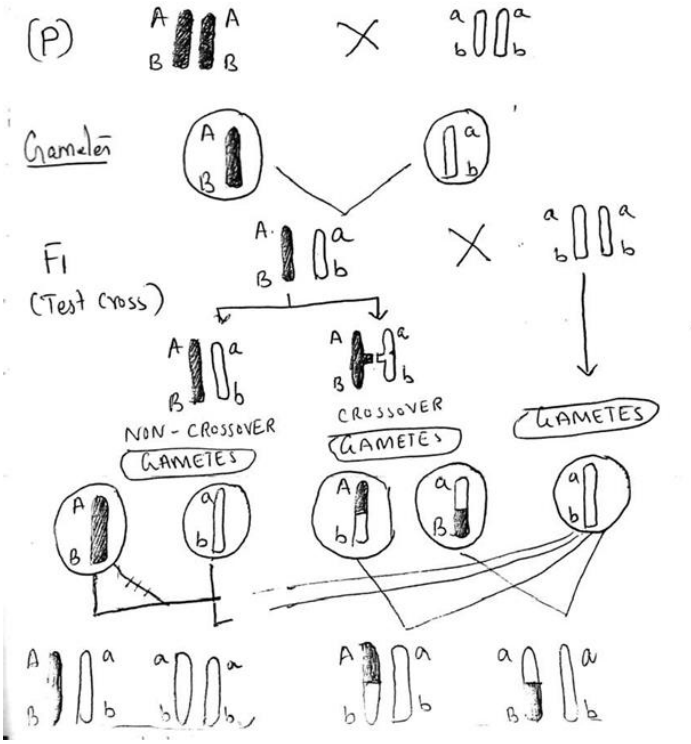
Genes > Chromosome

c) All the genes present on a chromoson together constitue a linkage group. such genes are called as linked genes.

d) The number of linkage groups of a species is equal to the haploid number of chromosome.

e) The linked genes remain together on a chromosomes and are inherited as a unit.

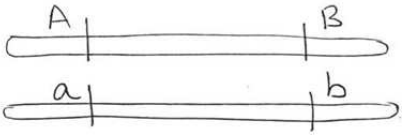
TEST CROSS SHOWING CROSS OVER AND NON - CROSS OVER GAMETES



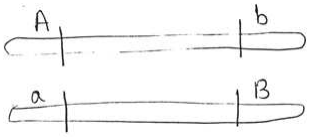
Arrangement of Linked Genes:

In heterozygotes having two pairs of linked genes are of two types. They are

a) C is - arrangemen: In this type, in the given pair of linked genes, two dominant allelas are located on one homologue of chromosomal pair and their recessive allele present on other homologous chromosome. Such heterozygotes are called C is - heterozygotes. Eg AB/ab.



b) Trans-arrangement: In this type, dominant allele of one pair and recessive allele of other pair are located on the same homologous chromosome. Such heterozygoes are called Trans-heterozygotes. Eg: Ab/aB.



TYPES OF LINKAGE

Linkage is of two types based on appearance and Non - appearance of new combination of charecters in the off spring.

i) Complete Linkage
ii) Incomplete Linkage

COMPLETE LINKAGE:

In this type, linked genes are transmitted together in parental combination for one or more generation. They donot separate from each other to form new character complete linkage is very rare.

Example:

In wild drosophila, the genes for grey body color is (B+) and vestigial wings (Vg) are linked. And another drosophila having black body (b) and long winged. (Vg+) are crossed.

Parental generation	**Grey vestigial**	**X**	**Balck long (Ebony)**
Genotype	$b^+ b^+$ Vg Vg	X	bb Vg^+ Vg^+
Gametes	(b^+) (Vg)		(b) (Vg^+)
F_1	b^+ b Vg^+ Vg	$\rightarrow$	Grey long

The F1 hybrids are test crossed with doubly recessive black vestigial wing flies C.B. Bridges discovered complete linkage.

Test Cross	b^1 b Vg^+ Vg F_1 Grey long	X	bb Vg Vg Black vestigial
Gametes	$(b^+$ Vg) (b $Vg^+)$		(b Vg)

PUNNETT SQUARE

	b Vg	Phenotype
b^+ Vg	b^+ b Vg Vg	Greyvestigial
b Vg^+	bb Vg^+ Vg	blacklong

Incomplete Linkage :

In majority of organism, the linkage is incompl,ete. New charectess appear in the off spring due to crossing over betwen homologous chromosome. Hutchinson discovered incomplete linkage on endosperm charecters in Maize.

Example:

For coupling Phase

- ✓ Coloured aleurono (C) is dominant
- ✓ Colourless aleurone (c) is recessive
- ✓ Full endosperm (S) is dominant
- ✓ Shrunken endosperm (s) is recessive

Parental	Coloured full		Colourless shrunken
Generation	C C S S	X	c c s s
Gametes	(C) (S)		(c) (s)
F_1 Generation Test cross	Cc Ss Cc Ss	X	coloured full c c s s

Phenotypic ratio of test cross progeny.

♀/♂	cs	Phenotype	Expected Test cross ratio	Observed data out of 450
CS	Cc Ss	Coloured full	100	180
Cs	Ccss	Coloured Shrunken	100	20
cS	ccSs	Colourless full	100	20
cs	ccss	ColourlessShrunken	100	180
			1:1:1:1	400

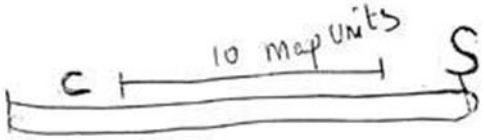
Linkage and Recombination frequency as for total population are.

Parental characters (non cross over) = $\frac{180}{180}$ x 100 = 90%

Non-parental characters (cross over) = $\frac{20+20}{400}$ x 100

1% RF = 1 cM = 1 map units = 10%

Chromosomal map



Example for repulsion phase

- **Parental generation :** Coloured shronken X colourless full

crenotype	CCss	X	c c S S
Gameter	(C) (s)		(c) (S)
F_1 Test cross	C c Ss Cc Sc	X	Coloured full c c s s

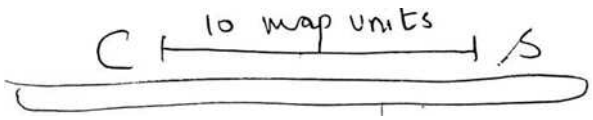
♀/♂	cs	Phenotype	Expected Test cross ratio	Observed data out of 450
CS	Cc Ss	Coloured full	100	20
Cs	Ccss	Coloured Shrunken	100	180
cS	ccSs	Colourless full	100	180
cs	ccss	ColourlessShrunken	100	20
			1:1:1:1	400

Linkage & Recombination frequencies as for total population are

Parental charector (non cross over) = $\frac{18+180}{400}$ x 100 = 90%

Non-parental charecters (cross over) = $\frac{20+20}{400}$ x100 = 10%

Coromosomal map:



The crossing over % converted into map distance which exprened as map units or morgan units (6,4) 1% crossing over = 1 map unit.

CROSSING OVER

- This was first discovered by T.H. Morgan and castle in 1912. When linked genes tend to get separated and make new combinations. This type of mechanism are called CROKSSING, OVER
- The salient features of crossing over are
  - crossing over is an alternative to linkage
  - It separates linked genes on the same.

Chromo some:

- They occur due to exchange of corresponding parts of non sister chromatids of homologous chromosome.
- Crossing over depends upon the distance between the genes.

MECHNISM OF CROSSING OVER

Crossing over occurs during meiosis, during pachytene stage of Meiotic prophase I.

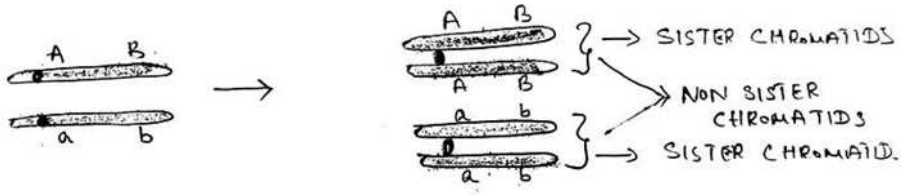
Meiosis → Prophase I → Pachytene stage

Crossing over occurs in the slage,

1. SYNAPSIS STAGE
2. CROSSING OVER STAGE

SYNAPSIS:

In this stage, homologous chromosome pair with each other during zygotene. Pairing occurs between paternal and maternal chromosomes and are twisted about each other. At this stage each chromosome has two chromatids attached to common centeromere.

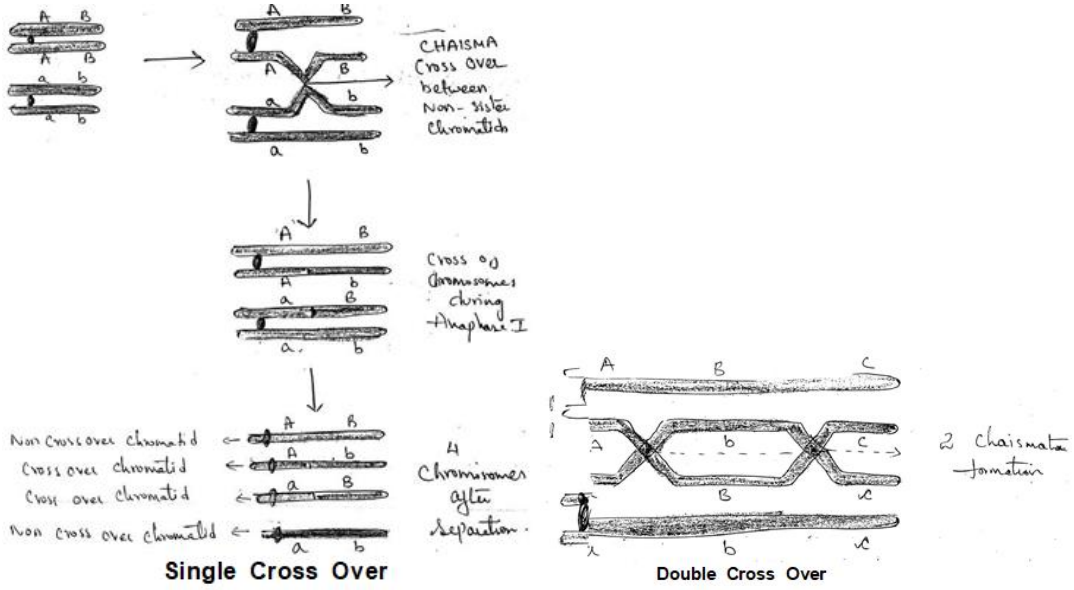


Crossing Over:

It occurs in pachztene stage of prophase I, in which parts of chromotids gets exchanged between Non sister chromatids. Important aspects of crossing over as follows.

1. Crossing overs occurs between non-sister chromatids.
2. Only two chromatids participate in crossing over.
3. Crossing over are identified by chiasmata formation, which are the points for crossing over
4. Nomber of chiasmata indicates the number of cross overs between the chromosomes.
5. The frequency of cross overs depends on the distance between the genes and length of chromosomes.

Chaisma cross over



CYTOLOGICAL EVIDENCES OF CROSSING OVER

Crossing over was proposed by T.H.Morgan in 1914, but it was clearly demonotrated by a cytologically in 1931. because okf following rease Homologous chromosomes exchange reciprocal segments of chromatids.

1. Chromosomes after exchange don't show morphologica differences.
2. These is no other cytological observation in crossing over except chiasmata.

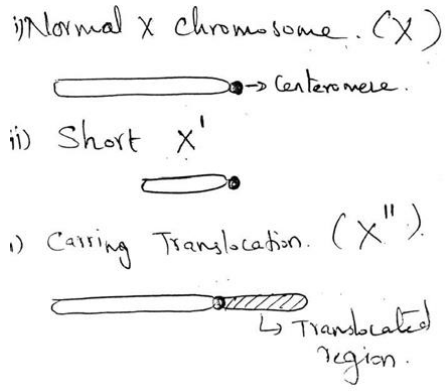
Chromosomes are connected by chiasmata, but it is not necessary that cliasmata should be associated with the crossing over.

Example for cytological evidence of crossing over :

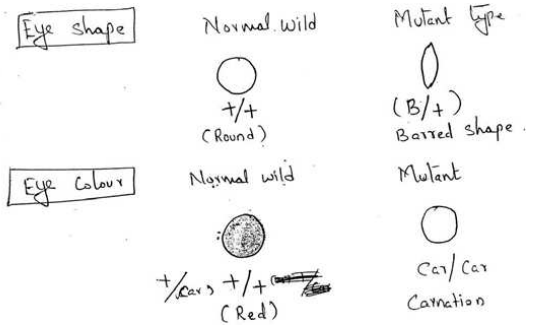
Curt stern (1931) in Drosophila.

- In 1931, C. Stern developed new drosophila female strain which has two different X- chromosomes.
- One X - chromosome found to be shortar and another has a small piece of chromosome IV attached, along with X-chromosome translation. This is called Broken X-chromosome. The shorter X chromosome is resered X and translcated X ch along.

Types of X – chromosome

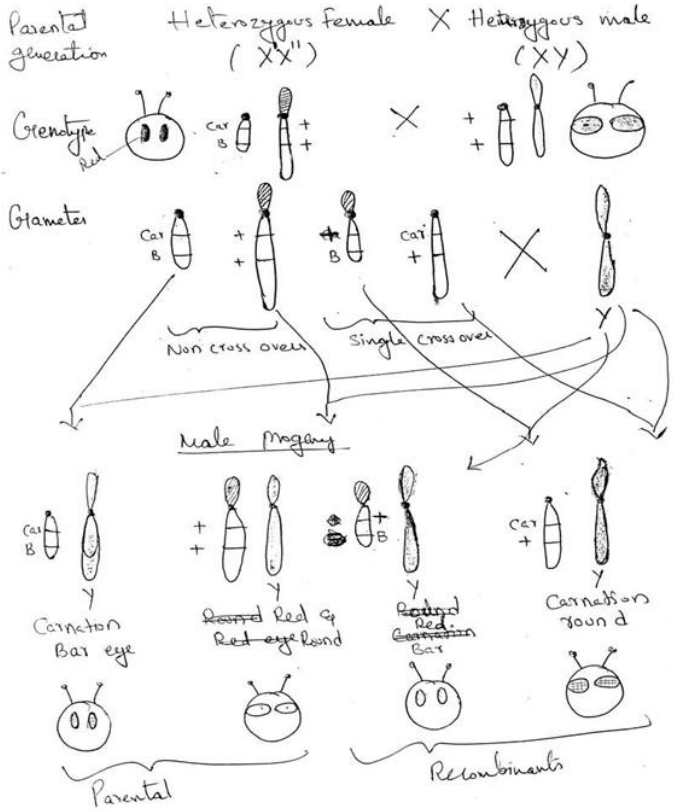


Local Under Study in drosophola



BB	- Produces	Barred shaped	eye	(BB/Bb)
cc	- Produces	Carnation eye	colour	(only cc)
bb	- Produces	round	eye shape	(only bb)
CC	- Produces	Red	eye colour	(CC/Cc)

- The receissive gene 'Car' produces carnadian eye color instead of Normal red eye color and the dominant gene 'bar' produces bar like eye insteed of normal oval eye.
- This experiment concluded that physical basis of recombination between linked genes is exchange of chromosomal segments between homologous chromosome by crossing over



P: C c Bb X c c bb

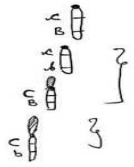
Gametor (CB) (Cb) (c) (b)
(cB) (cb)

Punneft square

	cb	Phenotype	Phenotype appearance
CB	CcBb	Red colourBar eye	
Cb	Ccbb	Red colourRound eye	
cB	ccBb	Carnation colourBar eye	
cb	ccbb	Carnation colourRound eye	

4 types of X - chromosomes are observed in all the four types of male flies.

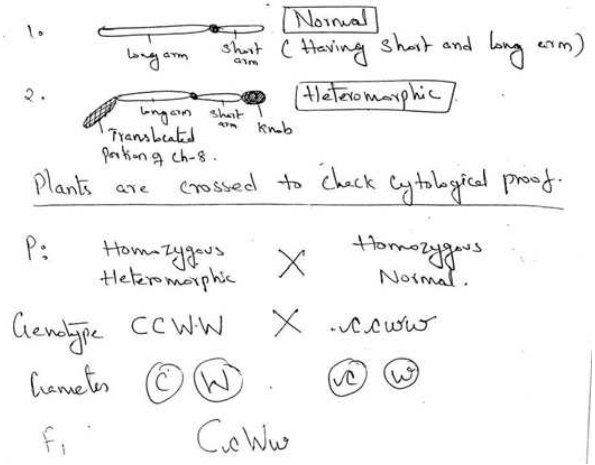
i) Carnation bar : c = car } Parental type
ii) Carnation round : recombinant type
iii) Red bar : Recombinant type
iv) Red round Parental type



Mc Clintock Experiment in Maize

- H.S. Creighton and Barbara Mcclintock in 1931 demonstrated crossing over in maize cytologically.
- The strain of corn had two morphologically distinct features on chromosome 9 which could be easily identified under microscope.

Types of chromosome 9.



RECOMBINATION REQUENCY MAP DISTANCE

Recombination Frequency (RF)

- Frequencies of crossing over also called as recombination frequency.
- RF between linked genes are estimated from test cross progeny. This procedure was developed by fishes.
- The four phenotypic classes.

- [AB],[(Ab], [aB], [ab] are assigned
- The value of RF (Z-value) is determined is follows.
- In case of coupling phase linkage.

$$Z = \frac{(a_2 \times a_3)}{(a_1 \times a_4)}$$

Where a_2 and a_3 are recombinant progency & a_1 and a_4 are parental.

g : (check the talbe from linkage)

$$Z = \frac{(20 \times 20)}{(180 \times 180)} = \frac{400}{32400} = 0.0123$$

Hence the RF value for this case is 0.0123.

In case of repulsion phase linkage.

$$Z = \frac{(a_1 \times a_4)}{(a_2 \times a_3)}$$

Where a_1 and a_4 are recombinant progency a_2 and a_3 are parental progency.

g : (check table from linkage)

$$Z = \frac{(20 \times 20)}{(180 \times 180)} = \frac{400}{32400}$$

$$= 0.0123$$

Hence the RF valuae for this case is 0.0123.

If checking the Z valuae table, the recentage recombination for loupling phase is 5.1 and for repulsion it is 7.0.

Percent Recombination table

Value of Z	Coupling phase	Repulsion phase
0.001	2.7	2.2
0.002	3.7	3.2
0.003	4.6	3.9
0.010	8.1	7.0
0.02	11.1	9.9
1.00	50.0	50.0

MAP DISTANCE / CHROMOSOMAL MAP

It is defined as the graphical representation of genes on the same chromosome showing the distance between different genes of linkage group.

Following steps are required to construct chromosomal maps.

1) The number of linkage groups are identified for the given species.
2) Determination of distance between the genes by cross over % of recombinants.
3) Cross over % is converted into map distance which is expressed in map units or centimorgan units (cM), i.e.

1% of crossing over = 1 map units

chromosomal map is also called as linkage map, genetic map.

These are two types of test crosses used determine the distance between the genes.

i) Two point test cross

ii) Three point test cross.

Point Test Cross (Dihybrid cross)

A maize variety having coloured alevrone and full endosperm (CCSS) crossed with excessive colourless aleurone and shronkan adosperm. (ccss)

Coloured full X colourless shronlean.

CC SS X ccss

(c) (s) (c) (s)

C c S s X Coloured full)

Test cross : C c Ss X ccss

(CS) (Cs) (cs)

(cS) (cs)

Four types of off springs are produced in the following ratio.

	cs	Phenotype	Number	Type
CS	CcSs	Coloured full	4032	Parental
Cs	Ccss	Coloured Shronkan	149	Recombinant
cS	ccSs	Colourless full	152	Recombinant
cs	ccss	Colourless shronken	4085	Parental
		1:1:1:1 Test cross	8418 Total	

$$\text{Parental frequency (Non cross over)} = \frac{4032 + 4085}{8418} \times 100$$

$$= \frac{8117}{8418} \times 100$$

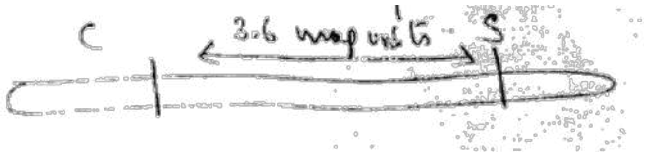
$$= 96.4\%$$

$$\text{Recombinant frequency (cross over)} = \frac{149 + 152}{8418} \times 100$$

$$= \frac{301}{8418} \times 100$$

$$= 3.6\%$$

$\therefore$ The chromosonal map is



Three Test Cross

coloured aleuron full endosporm & Starely endosparm		Colourless aloumn shnonken endospasm waxy endosperm
CCSSWW	X	ccssww
(C) (S) (W)	X	(c) (s) (w)
GEN Cs Sc Ww	=	(coloured, full, starely)
cross Cs Ss Ww	X	c c s s w w

The following data obtained C SW C Sw C sW Csw
c SW cSw csWcsw

henotype	Total No	No.	Native of cross over
	260	26.0	Non cross over
	240	24.0	Non cross over
	96	9.6	Single cross over b/w C-S
	104	10.4	Single cross over b/w C-S
	138	13.8	Single cross over b/w S-W
	142	14.2	Singl cross over b/w S-W
	12	1.2	Double over b/w C-S & S-W
	8	0.8	Double cross over b/w C-S & S-W
Total	1000	100	

i) Crossing over % b/w C-S

$$= \frac{\text{single crossover}(C-S) + \text{Double crossovers}}{\text{TotalNo.of offspring}} X100$$

$$= \frac{(96+104)+(12+8)}{1000} \times 100$$

$$= \frac{220}{1000} \times 100 = 22\% \text{ or } (22cM)$$

ii) Cross over % b/w S-W:

$$= \frac{\text{single cross over b/w } S-W + \text{Double cross overs}}{100 \text{ Total No.of off springs}} \times$$

$$= \frac{(138+142)+(12+8)}{1000} \times 100$$

$$= \frac{300}{1000} \times 100 = 30\% \text{ or } (30cM)$$

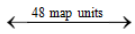
iii) Cross over % b/w C-W

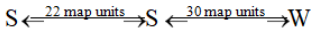
$$= \frac{\text{single cross over b/w C-S + single cross over b/w S-W}}{\text{Total No of off springs}} \times 100$$

$$= \frac{(96+104)+(138+142)}{1000} \times 100$$

$$= \frac{480}{1000} \times 100 = 48\% (18cM)$$

∴Distance b/w C-S = 22% (or) 22 map units
Distance b/w S-W = 30% (or) 30 map units
Distance b/w C-W = 48% (or) 48 map units
Chromosomal map represented as





4.2 NON - MENDELIAN INHERITANCE MATERNAL EFFECT

Shell Coiling In Snail

According to sturtevent, there are two strains of water snails (Limnaea peregra), that differ each other in the direction of coiling of shell i.e. one strain of the shell always coils to the left (sinistral), where as in other strain the shell always coils to the right (dextral)

Maternal Inheritance or Effect

The trait of female parent only transmitted to progeny. Hence there is no seggregation of genes in F2 generation such effect is called maternal effect.

1) The genes showing Non mendalian inheritance are present in nucleus or cytoplasm such genes are called cytogenes or Extra nuclear genes.
2) The coiling phenotype seen in the off spring is controlled by the genotype of Mother. Let's take the following crosses to understand better.

Example -1:

Female ( )	Male ( )
Dextral	Sinistral
Right coil	Left coil }Parent
DD	dd

D-Gene produces protein & makes dextral

Dd
(Dextral)

←Ddx Dd→ F_1

Dextral		**Dextral**	
	D	d	
D	DD	Dd	→F2
	Dextral	Dextral	
d	Dd	dd	
	Dextral	Dextral	

All off spring in F2 generation shows dextral right coil because mothers cytoplasm provides D-gene with D-protein.

Example - 2 :

Female ( )	**Male ( )**	
Sinistral	Dextral	Parental gen
Left Coil	Right coil	
dd	DD	
No Protain	(D) Gametes	

∴**sinistral**

Egg

Dd ___ F_1

Sinistral

F_1 X F_1

Dd X Dd

Sinistral Sinistral

Egg cytoplasm has D - protein hence all are dextral off springs

	D	d	
D	DD	Dd	
	Dextral	Dextral	F_2 gen
d	Dd	dd	
	Dextral	Dextral	

RECOMBINATION FREQUENCY & MAP DISTANCE

Recombination Frequency (RF):

- Frequencies of crossing over also called as recombination frequency.
- RF between linked genes are estimated from test cross progeny. This procedure was developed by Fishes.
- The four phenotypic classes.
- (AB), (Ab), (aB), (ab) are assigned as a1, a2, a3 and a4. The value of RF (Z-value) is determined as follows.

a) In case of coupling phase linkage.

$$Z = \frac{(a_2 \times a_3)}{(a_1 \times a_4)}$$

Where a_2 and a_3 are recombinant progency & a_1 and a_4 are parental. Eg: (check the table from linkage)

$$Z = \frac{(20 \times 20)}{(180 \times 180)} = \frac{400}{32400} = 0.0123$$

Hence the RF value for this case is 0.0123.

b) In case of Repulsion phase linkage

$$Z = \frac{(a_1 \times a_4)}{(a_2 \times a_3)}$$

Where a_1 and a_4 are recombinant progeny & a_2 and a_3 are parental progeny. Eg: (Check table from linkage)

$$Z = \frac{(20 \times 20)}{(180 \times 180)} = \frac{400}{32400} = 0.0123$$

Hence the RF value for this case is 0.0123.

By checking the Z value table, the percentage recombination for coupling phase is 8.1 and for repulsion it is 7.0.

Percent Recombination table

Value of Z	Coupling Phase	Repulsion phase
0.001	2.7	2.2
0.002	3.7	3.2
0.003	4.6	3.9
0.010	8.1	7.0
0.02	11.1	9.9
1.00	50.0	50.0

MAP DISTANCE / CHROMOSOMAL MAP

It is defined as the graphical representation of genes on the same chromosome showing the distance between different genes of linkage group.

Following steps are required to construct chromosomal maps.

1. The number of linkage groups are identified for the given species.
2. Determination of distance between the genes by cross over % of recombinants.
3. Cross over % is converted into map distance which is expressed in map units or centi Morgan units (cM), i.e.
   1% of crossing over = 1 map units
4. Chromosomal map is also called as linkage map, genetic map.
5. There are two types of test crosses used determine the distance between the genes.
   a. Two point test cross.
   b. Three point test cross.

Two Point Test Cross : (Dihybrid cross)

A maize variety having coloured aleurone and full endosperm (CCSS) crossed with recessive colorless aleurone and shronken endosperm. (ccss)

p: colored full X colorless shrunken

Genotype	CCSS	X	ccss
Ganuse	(C) (S)		(c) (s)
F_1 :	C c S s		(Colored full)

Fix Testcross : Cc Ss X cc ss

(Cs) (Cs) (cs)

(cS) (cs)

Four types of off springs are produced in the following ratio.

	cs	Phenotype	Number	Type
CS	CcSs	colored full	4032	Parental
Cs	Ccss	colored shrunken	149	Recombinant
cS	ccSs	colorless full	152	Recombinant
cs	ccss	colorless shrunken	4085	Parental
		1 : 1 : 1 : 1	8418	
		Test cross	Total	

Parental frequency (non cross over) $= \frac{4032+4085}{8418} x100$

$= \frac{8117}{8418} x100$

$= 96.4\%$

Recombinant frequency (cross over) $= \frac{149+152}{8418} x100$

$= \frac{301}{8418} x100$

$= 3.6\%$

∴The chromosomal map is 3.6 map units

Three Test Cross (Tri Hybrid Cross)

P : Colored aleuron full endosperm & Starchy endosperm	X	Colorless aleuron shrunken endosperm waxy endosperm.
CC SS WW	X	cc ss ww
(C) (S) (W)	X	(c) (s) (w)
Cc Ss Ww		(Colored, full, starchy)
Cc S s W w	X	c c s s w w

The following data obtained CSW CSw CsW Csw
cSW cSw csW csw

Phenotype	Total No.	Frequency	Nature of cross over
Colored full starchy CSW	260	26.0	Non cross over
Colorless shrunken wary csw	240	24.0	Non cross over
Colored shrunken wary CsW	96	9.6	Single cross over b/w C-S
Colorless full starchy	104	10.4	Single cross over b/w C-S
Colored full wary	138	13.8	Single cross over b/w S-W
Colorless shrunken starchy	142	14.2	Single cross over b/w S-W
Colored shrunken starchy	12	1.2	Double cross over b/w C-S & S-W

Colourless full waxy	8	0.8	Double cross over b/w C-S & S-W
Total	**1000**	**100**	

a) Crossing over % b/w C- S

$$= \frac{\text{single corssover (C – S)} + \text{Double crossovers}}{\text{Total no. of off spring}}$$

$$= \frac{(96+104)+(12+8)}{1000} \times 100$$

$$= \frac{220}{1000} \times 100 = 22\% \text{ or (22cM)}$$

ii) Cross over % b/w S-W :

$$= \frac{\text{single cross over b / w S W} + \text{Doublecross overs}}{\text{Totalno.of off springs}} \times 100$$

$$= \frac{(138+142)+(12+8)}{1000} \times 100$$

$$= \frac{300}{1000} \times 100 = 30\% \text{ or (30cM)}$$

iii) Corss over % b/w C-W

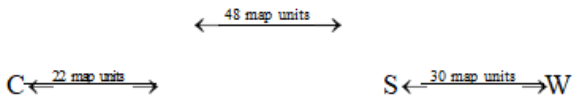
$$= \frac{\text{single crossover b / w C S} + \text{single b / w crossover} - \text{S W}}{\text{Total No: of offsprings}} \times 100$$

$$= \frac{(96+104)+(138+142)}{1000} \times 100$$

$$= \frac{480}{1000} \times 100 = 48\% \text{ (48c M)}$$

∴ Distance b/w C-S = 22% (or) 22 map units
Distance b/w S-W = 30% (or) 30 map units
Distance b/w C-W = 48% (or) 48 map units

Chromosomal map represented as



VARIEGATION IN LEAVES OF MIRABILUS JALAPA (4 O CLOCK PLANT)

Plastid inheritance, i.e. plastids are cytoplasmic organelles in plant cells. There are 3 types of cells present in mirabilis Jalapa namely.

1. i) Cells with chloroplast Green (G)
2. ii) Cells with leucoplast Pala green (p)
3. iii) Cells with chloroplasts leucoplasts & Variegation (v)

Example - 1:

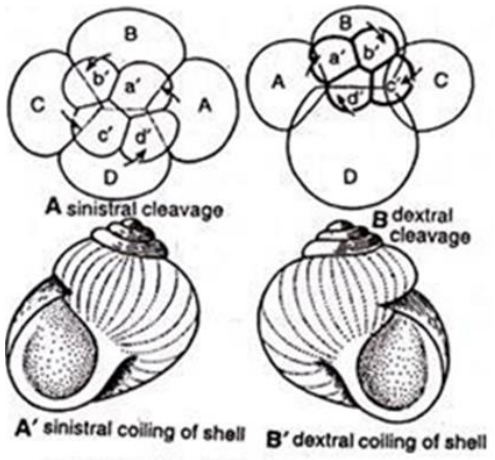
G - Green leaf

P - Pale leaf

v - Variegation leaf

Shell coiling in Snail and Maternal effects

When a female gamete fuses with a male gamete, an embryo is created. The female gamete supplies the cytoplasm for the growing embryo and is physically bigger than the male gamete in the great majority of species. Factors produced by the female's nuclear genes are found in this cytoplasm. These elements could affect the growing embryo in particular ways. All species' mitochondria and plant species' chloroplasts are likewise derived from the female cytoplasm. These two organelles regulate certain characteristics in the progeny and contain DNA. Phenotypes that exhibit a maternal impact are those that are regulated by nuclear factors present in the female's cytoplasm. Maternal inheritance is present in those traits that are regulated by organelle genes.



Coiling snail shells are a typical trait that shows maternal impact. The mother's genotype determines the coiling phenotype that the kids display. The outcomes of the following crosses between pure line snails were observed. It is customary to offer the female first.

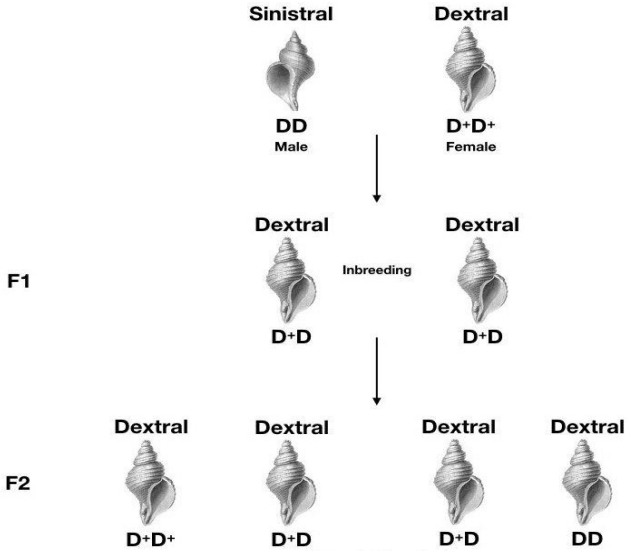
	Cross 1	Cross 2
Parents	Right coil x Left coil	Left coil x Right coil
F1	All right coil	All left coil
F2	All right coil	All right coil
F3	3 right coil : 1 left coil	3 right coil : 1 left coil

On the surface, these findings seem to contradict Mendel's rules. First, the two crosses have different F1 phenotypes. The results of reciprocal crossing

complementary crosses in which the male and female phenotypes are reversed in the original parental cross were comparable in previous tests, but in this one, it seems that the female controls the phenotypic. However, since the F1 animals that were left- and right-coiled had all-right offspring, the F2 seems to defy this theory. Additionally, the F3 generation exhibits the 3:1 Mendelian ratio rather than the F2.

The offsprings whose mothers are either homozygous or heterozygous for right coiling are right coilers even if they are homozygous for sinistrality (left coiling). In the same way offspring of left coiling mother are left coilers even if they carry dominant genes for right coiling.

The F2 females of either cross (right coiler) when mated with males of any genotype produce at an average right coilers and left coilers in 3: 1 ratio. But 3: 1 ratio appears in F3 and not in F2. If F2 males are mated with homozygous right coiling females, there is no segregation and all their progenies are right coilers, but if they are mated with homozygous left coiling female's only left coilers are produced.



Carl Correns (1908)

Put forth in cytoplasmic inheritance Mirabilus Jalapa for O' clock plant

i) When green branches are used as female source, & when crossed with male source of green, pale or variegation branches, the off spring produced are green branches, because green chloroplast is present in egg cytoplasm & males doesn't contribute cytoplasm. Thus it is maternal plastid inheritance.

ii) When female pale branch crossed with either of green, pale or variegation male branch, the progeny appeared is pale branches only.

iii) When variegated branches are used as female source, all the three types of plastids are present in the female parent. Therefore, all the 3, chlorophyll, leucoplast & chromoplast appears in the progeny

∴ All 3 branches green, palae & variegation plants would be obtained.

1) Branch (Green) X Branch (Green, pale or variegation)

↓

F_1 is green plants

2) Branch (pale) X Branch (Green, pale variegation)

↓

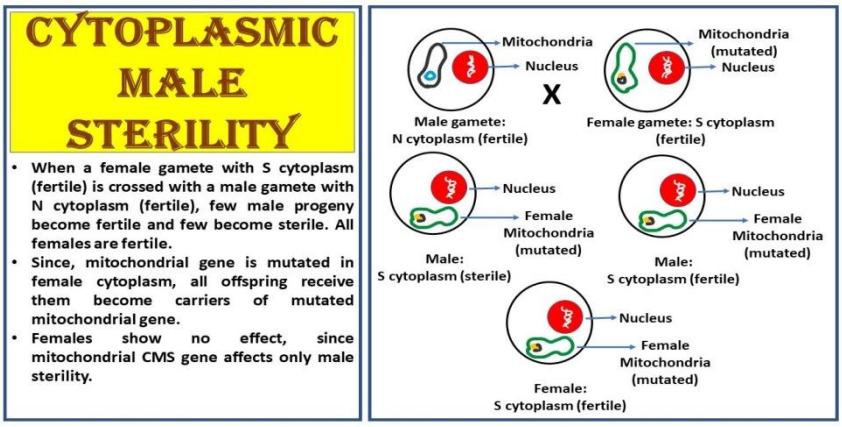
F_1 is pale plants only

3. (Branch variegated) X Branch (Green pale, variegated)

↓

F_1 is green, pale & variegated plants.

4.3 Cytoplasmic male sterility in maize



In hermaphrodite species, cytoplasmic male sterility refers to either complete or partial male sterility due to certain nuclear and mitochondrial interactions. The inability to generate viable anthers, pollen, or male gametes is known as male sterility. Gynodioecious communities—populations with coexisting fully functional hermaphrodites and male-sterile hermaphrodites—are caused by such male sterility in hermaphrodite populations.

As the term suggests, extranuclear genetic control—that is, control over the mitochondrial or plastid genomes—is responsible for cytoplasmic male sterility. Male sterility is transmitted maternally, demonstrating non-Mendelian inheritance. Cytoplasm often comes in two varieties: aberrant S (sterile) and N (normal). There are reciprocal distinctions among these categories.

History

Joseph Gottlieb Kölreuter was the first to document male sterility in plants. In the 18th century, he reported on anther abortion within species and specific hybrids. Cytoplasmic male sterility (CMS) is mostly found in angiosperms and has been identified in more than 140 angiosperm species.

CMS has also been identified in one animal species so far, Physa acuta, a fresh water snail. There is strong evidence for gynodioecy and CMS to be a transitionary step between hermaphrodites and separated sexes.

Male sterility is more prevalent than female sterility. This could be because the male sporophyte and gametophyte are less protected from the environment than the ovule and embryo sac. Male-sterile plants can set seed and propagate. Female-sterile plants cannot develop seeds and will not propagate..

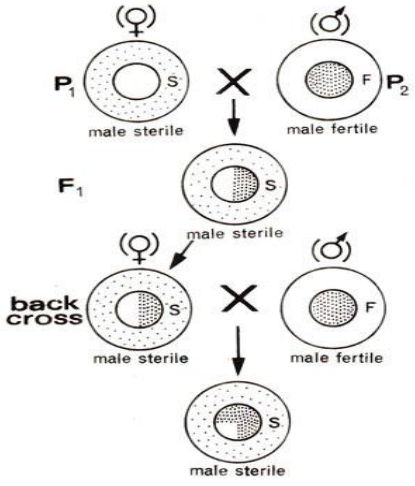
Male sterility is easy to detect because a large number of pollen grains are produced in male fertile plants. Pollen grains can be assayed through staining techniques (carmine, lactophenol or iodine).

Genetic sterility

Nuclear genes may be able to restore fertility, even if CMS is governed by an extra nuclear genome. Cytoplasmic–genetic male sterility occurs when nuclear restoration of fertility genes is possible for a CMS system in any crop; the sterility is characterized by the effect of both cytoplasmic (inherited from the mother) and nuclear (inherited from the father) genes. Additionally, certain genes that are different from hereditary male sterility genes are known as restorers of fertility (Rf) genes. Without the sterile cytoplasm, the Rf genes do not express themselves. To restore fertility in S cytoplasm that results in sterility, Rf genes are necessary. As a result, plants with N cytoplasm are fertile, whereas those with S cytoplasm and genotype Rf-are fertile, and those with rfrf are exclusively male sterile.

N cytoplasm with Rfrf is ideal for consistent fertility since Rf mutations, or mutations to rf or no fertility restoration, are common in these systems.

Because it is easy to manipulate sterility expression by adjusting the gene–cytoplasm combinations in any chosen genotype, cytoplasmic–genetic male sterility systems are often used in agricultural plants for hybrid breeding. By using these male sterility systems, cross-pollinated species can avoid emasculation, which promotes cross-breeding and results in only hybrid seeds in the wild.



4.4 The Poky Strain in Neurospora Crassa:

- Best Example for Mitochondrial inheritance in fungus Neurospora Crassa. In this fungi, some mitochondrial enzymes are deficient.
- Mitochondrial enzymes like cytochrome a,b and c. usually represents as cyt a, cyt b, cyt c. These enzymes are very important for electron transport which is necessary for oxidative phosphoxytation. i.e. ATP production.
- But in poky strains, these mitocondrial enzymes such as cyt a & cyt b is deficient but but c is present. This type of strain are called Normal poky strain. The pokyness is clue to absence of cyt a and b Hence poky strais grow slow.
- These are several mutant strains of poky strains which may differ in deficiency of cyt a, cyt b.
- i.e. one mutant poky strain is deficient in only cyt a while other is deficient in cyt b. These strains are called as mutant poky strain, they grow slow.

Example for Non-Mendelian Inheritance:

1. Normal poky X Mutant poky

F_1 generation Normal poky

2) Mutant Poky X Normal Poky

Mutant Poky in F_1 generation

- Hence male gametes in Neurospora contributes negligible amount of cytoplasm just like higher animals & plants.
- Only female cytoplasm is inherited in progeny i.e.
- The factor for pokyness is resided in the female cytoplasm.
- This non-mendelian uniparental inheritance suggested that the cytoplasm of female parent was important

Poky Mutant of Neurospora crassa

↓

Absence of cyt a (or) cyt b (or) both which are expressed in Mitochondrial DNA

↓

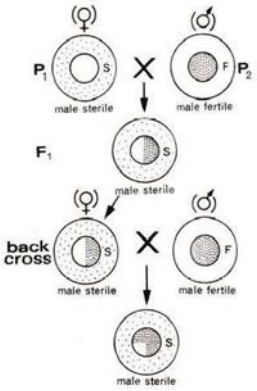
Petite Phenotype

↓

Grow very slow (Pokyness)

LIFE CYCLE OF NEUROSPORA CRASSA :

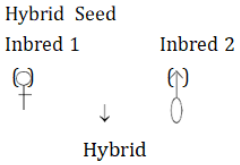
Cytoplasmic Male Sterility in Maize (Zea Mays) Introduction:



- Cytoplasmic male steritity (CMS) is the failure of plants to produce functional anthers, pollen or male gametic
- It shows Non - Mendelian inheritance, with male sterility inherited maternally. 2 types of cytoplasm.
    1. N (Normal)
    2. S (aberrant) is sterile
- CMS is important in hybrid maize production. It is 1st discovered in Texas i.e. hence its called as CMS-T. Later CMS - C (Charrua) & CMS - S (USDA) United State department of Agriculture.

Why Male sterility?

1. Reduce the cost of hybrid seed production.
2. Production of large scale F1 seeds.
3. Seed up the hybridization programme
4. Commercial exploitation of hybrid vigour.



Different ways for Male steritity

1. Emasculation (Detasselling)
2. CMS
3. Genetic male sterility (ms 1 to ms 52)

i) CMS - T → T - (Motation in Urf 13 gene)

↓

Important for electron transfer, ATP formation

↓

Male sterility

ii) CMS - C - Mutation in 3 genes atp 6, atp 9 & cos 11

iii) CMS - S - Mutation with orf 355 - orf 77

Restoration of fertility

If Nucleus has rf rf gene then 'S' cytoplasm cannot be restored to N cytoplasm.

4.5 CHLOROPLAST INHERITANCE IN CHLAMYDOMONAS REINHARDII

Baur and correns discovered non - Mendelian inheritance in plants in 1908. In 1950's chiba et e al, discovered that mitochandria and chloroplasts had their own genomes.

Homoplasmy & Heteroplasny

The number of copies of organelle genous (mitochondria or chloroplast) per organelle can vary from one to many.

- A cell or organisms in which all copies of an organelle gene are same is called homoplasmic & said to exhibit homoplasmy.
- A cell or organism in which not all copies of an organella gene are same is called Heteroplasmic & said to exhibit Heteroplasmy.

Diagrematic Representation of Homo - Heteroplasmy Cells

Diagram: Homoplasmic cells have same gene type Variegated } Hetermplasma cell

Diagram: Contain many alleles

Explain chloroplast inheritance of leaf - color phenotypes.

i) Egg cell ( ) X pollen ( )

If contains chloroplast / lerekoplast or variegated, H is passed into the F1 progency. This is beacause, Mother cytoplasm is contributed to progency not father's cytoplasm.

A chloroplast Mutant in Chlomydomonas sheptmycin Resistance

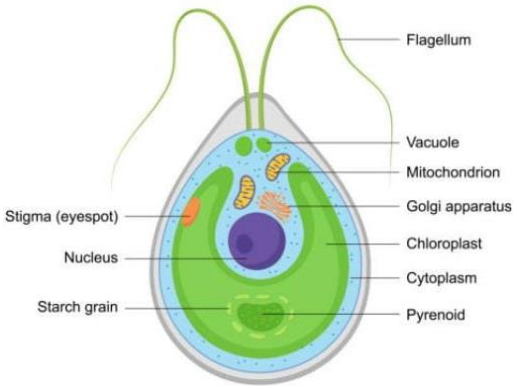
- The 1st Motation in chlanydomonas was discovered by ruth sagar (1954) and confers sheptomycin redistance (SmR) During mating of sheptomyin resistant (SmR) female clamydomonos with streptomyin sensitive (Sms) allele, the F1 gen observed was stnR, this is beguse matexnal inheritance observed in progency.

Diagram:

- Always Mt+ (mating type) cells one capable of mating while mt- are incapable.
- In this case, mother smR when crossed with Sm3 progeny observed were Sm^R
- In this example, Mother is mt^+ & sm^5 when crossed with father mt^- & sm^R the F_1 gen observed where mt^+ sm^5.
- Cytoplasmic inheritance is inherited from mother.

STRUCTURE OF CHLAMYDOMONAS REINIHARDH

CHLAMYDOMONAS



Mutant Chlamydomonas

1. $\frac{Mt^{+}}{Sm^{R}} \left| \frac{Mt^{-}}{Sm^{s}} \right. \rightarrow figen \frac{Mt+}{Sm^{K}}$
2. $\frac{Mt^{-}}{Sm^{R}} \left| \frac{Mt^{+}}{Sm^{s}} \right. \rightarrow Figen \frac{Mt+}{Sm^{R}}$
3. $\frac{Mt^{+}}{Sm^{s}} \left| \frac{Mt^{-}}{Sm^{R}} \right. \rightarrow Figen \frac{Mt^{p}}{Sm^{s}}$
4. $\frac{Mt^{+}}{Sm^{s}} \left| \frac{Mt^{-}}{Sm^{R}} \right. \rightarrow Figer \frac{Mt+}{Sm^{s}}$

4.6 HARDY - WEINBERG EQUIBRIUM (HWE):

1. The hardy - weinberg equilibrium states that, it is a principle where the genetic variation in a population will remain constant from one generation to the next in the absence of disturbing factors.
2. When mating is random in a large population with no distruptive circumstances, the law predicts that both genotype and allele frequency will remain constant, because they are in equilibrium.
3. The Hardy-weinberg equilibrium (HWB) can be disturbed by many forces such as.
   - **a)** Mutation
   - **b)** Natural selection
   - **c)** Non-random mating
   - **d)** Genetic drift
   - **e)** Gene flow

Mutation: Distropts allele frequencies.
Natural Selection: Distrupts gene frequencies.
Non-Random mating: Also distropts gene frequencies.
Genetic drift: Distropts allele frequencies.
Gene Flow: Distropts allele frequencies.

Because of all these distroptive factors, Hardy - Weinberg equilibrium rarely occurs in reality.

Hardy - Weinberg Principle

Parental generation

Phenotype	YY	Yg	yy
Genatypic frequency	0.49	0.42	0.09
No. of Individuals(500) }	245	210	45

No of alleles Y : 245 + 245 y: 45 + 45
(Total : 1000) = 490 + 210 : 90 + 210
= 700 = 300

Allelic frequency } $\frac{700}{700+30}$ $\frac{300}{700+300}$

= 0.7 = 0.3
= p = q

Hardy weinberg Analysis for genotype frequency.

	p(0.7)	q (0.3)
	Y	y
0.7	YY	Yy
P Y	$P^2 = 0.49$	pq = 0.21
0.3q y	Yy pq=0.21	yy $q^2 = 0.09$

$p^2 + 2pq + q^2 = 1$

$0.49 + 2(0.21) + 0.09 = 1$

Where p^2 = Homozygous dominant genotype
2pq = Heterozygous genotype
q^2 = Homozygous recessive genotype

Program-1:

You have sampled a population in which you know that the percentage of the homozygous receissive genotype (aa) is 36%, using this percentage calculate the following.

i) The genotypic frequency of "aa"?
Ans: 36%

ii) The allelic frequency of "a"?

Ans: The frequency of aa = 36%
i.e., q2 = 0.36
then q = 0.6
allelic frequency of a = 60%

iii) The allelic grequency of "A"? p + q = 1, q = 0.6
p = 1 - 0.6
p = 0.4
frequency of A is 40%

iv) What is the frequency of AA and Aa?
AA = p2 (p = 0.4)
= 0.4 0.4
= 0.16
frequency of AA is 16% Aa = 2 pq
= 2 0.4 0.6
= 0.48
frequency of Aa is 48%

v) The frequencies of 2 possible phenotype if "A" is dominant over 'a'.
Ans: If A is dominant,
Genotype will be AA or Aa
AA Aa
Frequency = 16% + 48%

Problem -2:

A very large population of randomly mating laboratory nice contains 35% white mice. White color is caused by Homozygous sucessive gemotype (aa). Calculate alletic & genotypic frequencies?

Sol:

Homozygous recessive genotype = aa
According to alletic frequency formula
p + q = 1, where q = a
∴q^2 aa
q^2 0.35 (35x 100)
Hence q= √0.35
q=0.59

p + q=1
p 1 q
p 1 0.59
=0.41

According to genotypic frequency

$p^2 + 2pq + q^2 = 1$

Where (i) p = 0.41

$p^2 = 0.17 \rightarrow (1)$

ii) $2pq = 2 \times 0.41 \times 0.59$

$= 0.48 \rightarrow (2)$

iii) $q^2 = 0.59 \times 0.59$

$= 0.35 \rightarrow (3)$

$p^2 + 2pq + q^2 = 1$

$0.17 + 0.48 + 0.35 = 1$

Hence the given observation satisties Hardy weinberg equilibrium.