

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, Matthias 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 division
- ▲ 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 Anton 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  $\mu\text{m}$  ( $1\mu\text{m}= 10^{-9}\text{m}$ ) in diameter. The smallest bacteria are about 100 to 200 nm ( $1\mu\text{m} = 10^{-12}\text{m}$ ) in diameter. Sizes of some common bacteria are shown below:

Shape	Arrangement	Examples
Coccus (Round shaped)	Monococcus	Micrococcus spp
	Diplococcus	Streptococcus pneumoniae
	Streptococcus(Chains)	Streptococcus mutans
	Staphylococcus(Bunches)	Staphylococcus aureus
Bacillus (Rod shaped)	Monobacilli	Pseudomonas aroginosa
	Streptobacilli	Bacillus anthracis
	Irregular bacilli	Mycobacterium



		tuberculosis
Spirilla (Spiral shaped)	Single spirals	Treponema pallidum
Comma Shaped	Single cells	Vibrio cholera
Crescent 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

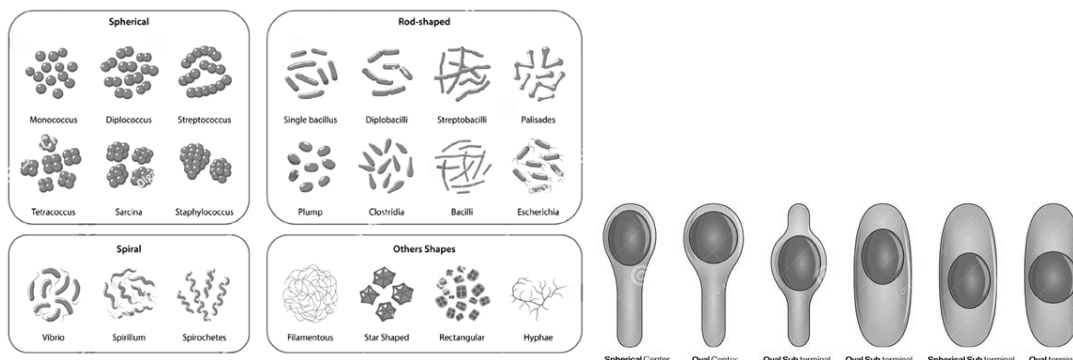


Fig. : Shapes of common bacteria

Fig. : Types of endospores

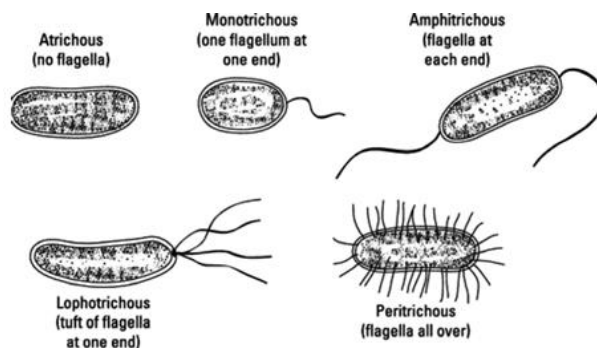


Fig. : Types of bacteria based on presence of flagella

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 ability 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 symmetry shown 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.

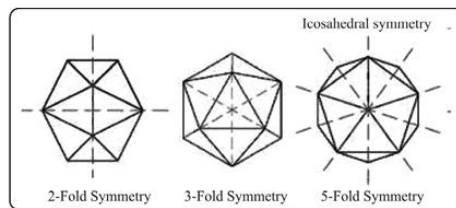
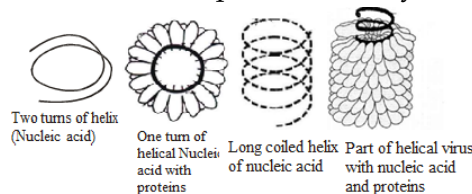
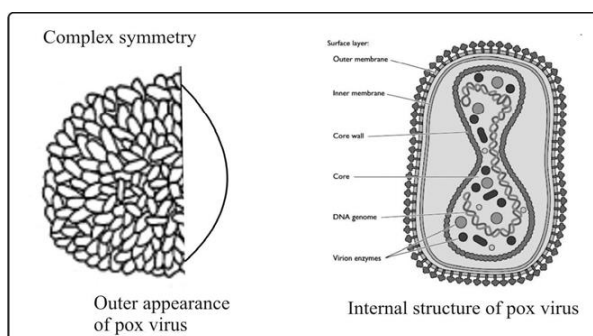


Fig. : Types of viral symmetry

- ▲ **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 membrane. 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 saprophytic 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 called hyphae (Singular - hypha). A hypha is usually 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 hyphae may be branched or unbranched. The hyphal wall consists of microfibrils 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 cytoplasm. The sub-cellular structures are supported and organized micro-tubules and endoplasmic reticulum.

Some common types of fungal cells are shown in Figure.



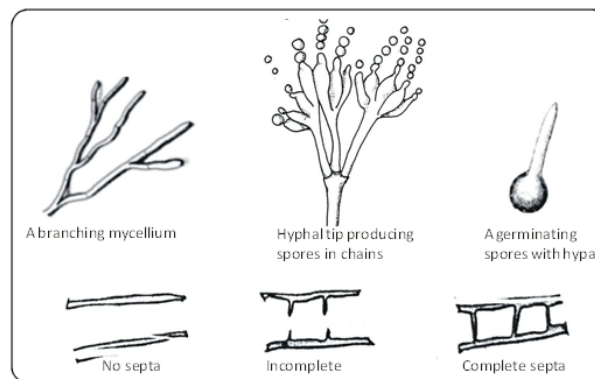


Fig. : Types of Fungal cells

### 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.

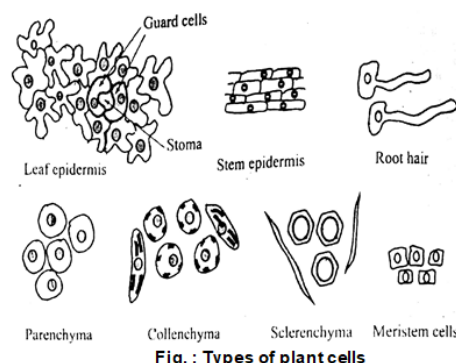


Fig. : Types of plant cells

**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 striated 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.

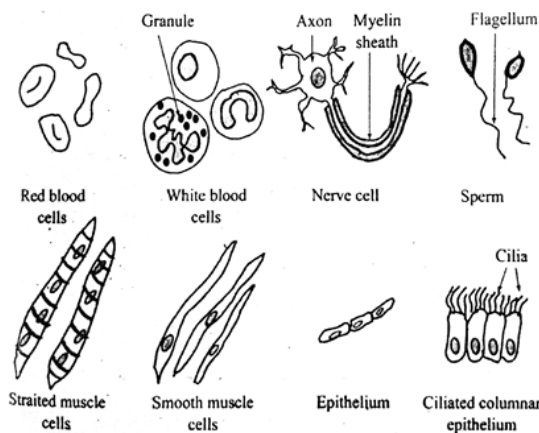


Fig.: Types of animal cells

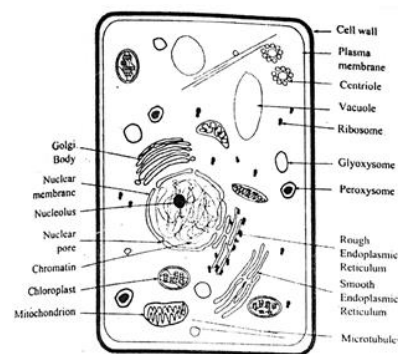


Fig.: Structure of plant cell

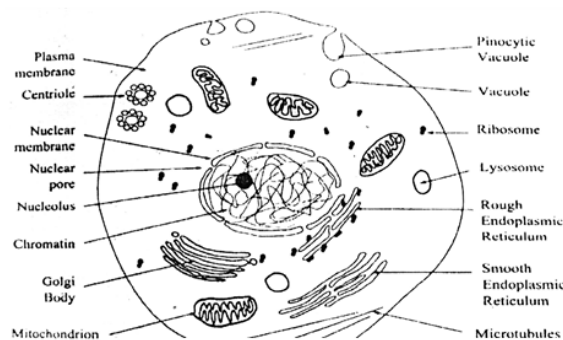


Fig.: Structure of an animal cell

## 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 termed prokaryotic (karyon means nucleus). In contrast, cells that have a nucleus are considered complex and evolved, and termed eukaryotic.

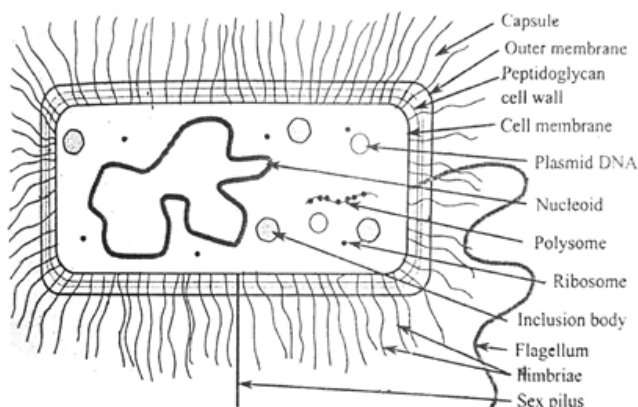
Prokaryotes are diverse in both size and shapes. They vary in size ranging from 0.1-0.2 $\mu\text{m}$  to 50 $\mu\text{m}$ . The smallest prokaryote is PPLO -'Pleuro Pneumonia like Organism', belonging to the Mycoplasma group whereas the largest prokaryote is *Thiomargarita namibiensis*. A prokaryotic cell is the unit of structure of prokaryotes, which includes Archaeobacteria, 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

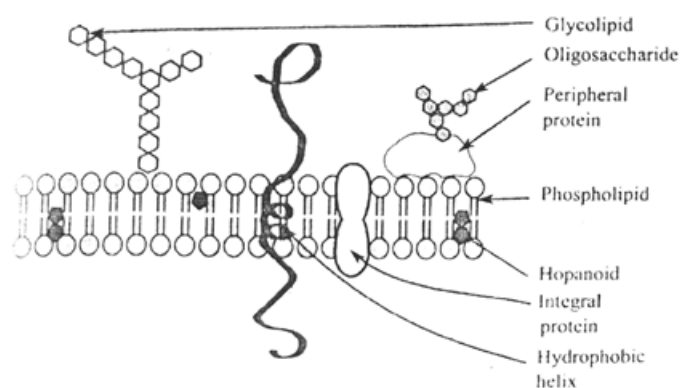
- (i) nucleoid
- (ii) ribosomes
- (iii) plasmids
- (iv) inclusion bodies
- (v) molecular chaperones
- (vi) thylakoids



### 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 by 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 respiratory 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 chromosomes.
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-exist 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, if 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 called 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, while ColE2 prevents DNA synthesis.
4. Degradative plasmids, which encode enzymes that can metabolize synthetic chemicals or xenobiotics such as salicylic acid, naphthalene and octane.



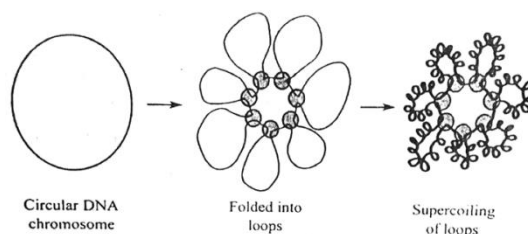
5. Metabolic plasmids, which encode enzymes for metabolic activities. For example, the Ti plasmid of *Agrobacterium tumefaciens* produces enzymes for the metabolism of opines. Other plasmids encode enzymes for denitrification activity in *Alcaligenes* spp., nitrogen fixation by *Rhizobium trifoli*, and for utilization of simple compounds like sucrose, lactose, and urea.
6. Virulence plasmids, which produce toxins that can harm other living 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 nucleoid 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 complexes 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 of *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 cell gluing them together. It is composed of pectin 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 cytoplasm 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 up to 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 cell and is made up of Phospholipids (20-30%) & protein (60-70%).

**Structure:**

- The plasma membrane is also called as cell membrane/cytoplasmic membrane.
- ▲ Singer and Nicholson in 1972 the 'Fluid Mosaic Model' of plasma membrane. This model is well accepted.
  - ▲ According to this model, the plasma membrane is a 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 maintained hydrophilic in nature whereas the internal part is hydrophobic in nature.
  - ▲ Plasma membrane is reflexed the non-polar (indulge in water) in which is maintained hydrophobic this condition is called as Amphiphatic.
  - ▲ Hopanoids are steroids like substances which provides stability for plasma membrane permits the components and nutrients to move laterally.
  - ▲ It is the site of generation of proton motive force. The force that drives ATP synthesis and also flagellar 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:** Carries additional molecules along with main molecules in the same directions.
  3. **ANTIORTER:** Transport of one molecule in one direction and other in opposite direction helps to maintain osmotic balance.

**Function:**

- ▲ The organic & inorganic nutrients are transported by permeases which are present in plasma membrane.
- ▲ It consists of enzymes required for biosynthesis pathway that synthesize components of cell wall such as
  - ▲ PGL, teichoic acid, phospholipids LPS etc.
  - ▲ It possess the attachment site for bacterial chromosome & plasmid DNA.
  - ▲ The inner membrane invaginates to form mesosomes as a site for Respiratory activities.
- ▲ It provides permeability barriers and 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.

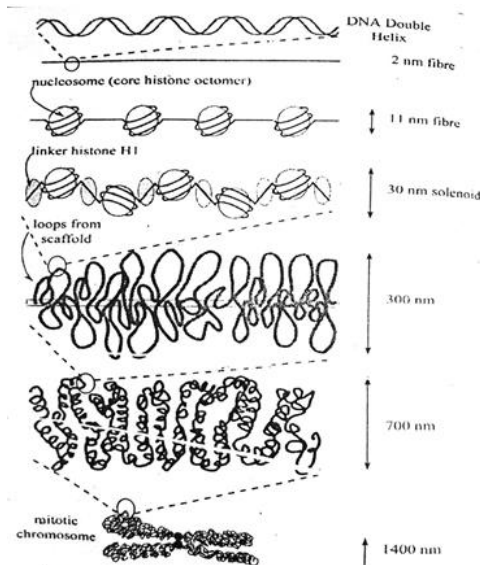


Fig. : Structure of eukaryotic chromatin

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.

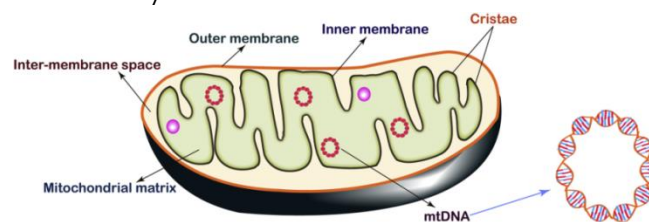
- ▲ **History:** Mitochondria was first do observed by Kollikes in 1850 as granular structures. Benda coined the term Mitochondria.
- ▲ **Distribution (or) Location:** Mitochondria has uniform distribution in cytoplasm.

### 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 urchin contains 1,40,000 to 1,50,000 etc.
- 2) **Shape:** The mitochondria may be filamentous or granular in nature and may change from one form to other. Depending on the physiological conditions of the cells. Thus they may be vesicular, ring or round shape.
- 3) **Size:** Normally mitochondria vary in size 0.5  $\mu\text{m}$  to 2.0  $\mu\text{m}$  sometimes their length may reach up to 7 $\mu\text{m}$ .
- 4) **Structure:** Mitochondria have two membranes. The membranes are phospholipid bilayer but inner and outer membrane are distinct 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 porins which allows, free exchange of molecules (nutrients directly into the cytosol).
- 2) **Inter membrane space:** Inter membrane space is between outer and 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 synthesis
  - ▲ Specific transporter proteins
 Inner membrane is site for production of ATP.
- 4) **Cristae:** Inner membrane folds into number of folding called cristae.
- 5) **Matrix:** The space formed by the inner membrane in folding 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/Mt 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 oxidized into  $\text{CO}_2$  and  $\text{H}_2\text{O}$ .
- ▲ During the biological oxidation of carbohydrates & fats large amount of energy is released which is utilized 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) thermogenesis. Biosynthesis (or) anabolic activities and accumulation of  $\text{Ca}^{2+}$  & phosphate in animal.
- ▲ They also involves in Glycolysis, Krebs cycle, oxidative decarboxylation 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 dysfunctions and 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 utilized by all living organisms to perform various life activities. The chloroplast is green in color 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, spherical, oval (or) club shapes.
- ▲ **Size:** The size of chloroplast various from species to species & generally measure 2-3  $\mu\text{m}$  in thickness & 5-10  $\mu\text{m}$  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 possess the layers they are  
1) Outer membrane      2) Inner membrane      3) Inter membrane space  
they contain 1-2% of total protein of chloroplast.



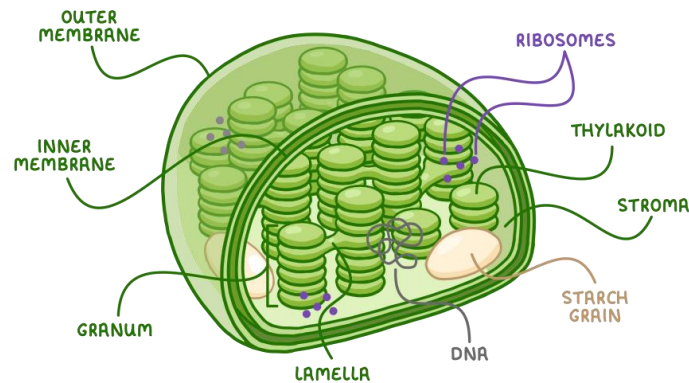


- ▲ **Stroma (or) Matrix:** It is also called as Matrix. It is maintained as a kind of jelly, fluid phase surround's the thylakoid the stroma contain Ribosomes & DNA which is involved in synthesis of structural proteins such as sugar, starch, fatty acid. They contain 50% of total protein of chloroplast
- ▲ **Thylakoids:** Thylakoids are sack like membrane organelles which is stack like a file of coins forming a structure called Grana. The number of thylakoids per grana varies from 1 to 50 or more. They form a network with in grana by lamella or inter granal fred. Within 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 energies.
- ◆ Palisade cells of leaves possess 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 colorless bodies called Photo Plastids. They are 3 types of plastids.

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

## 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 giant central vacuole.

The material seen in the vacuole is called as Sap. Cell sap contain salts, sugars, protein etc.

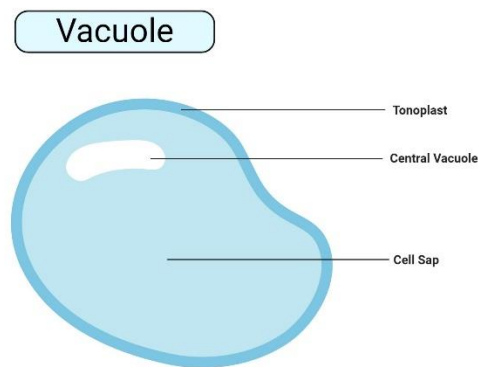


Figure: Vacuole, image Copyright © Sagar Aryal, www.microbenotes.com

### 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 Porter.
- ◆ **Occurrence:** The occurrence 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) cyto cavity 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 membrane (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 Ribosome 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

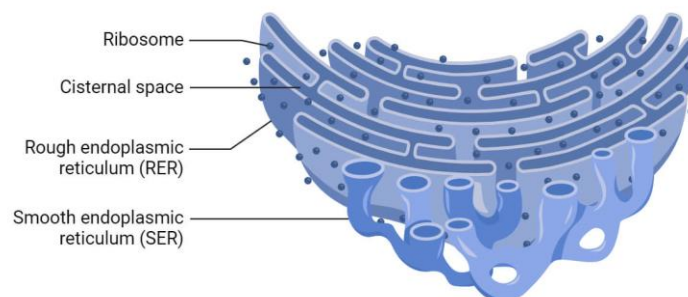
**Rough E.R:**

- ◆ 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 (80 S = 60S +40s) occurs in Rough E.R

**Functions:**

It provides mechanical support to the cell internally by forming ultra-structural skeletal frame work.

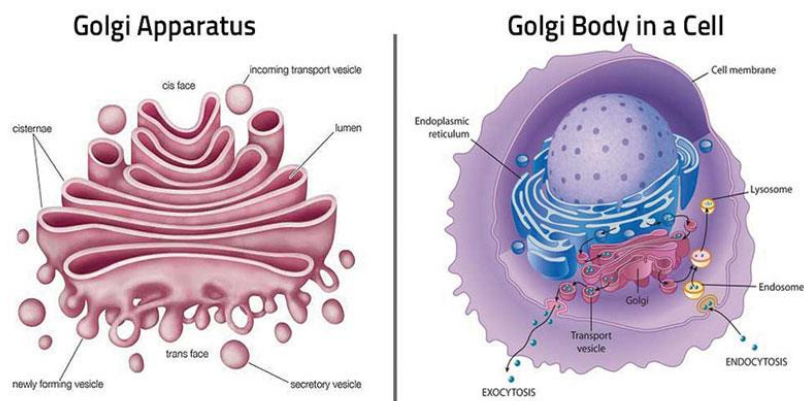
- ◆ It involves in transport of proteins.
- ◆ It acts as supportive structure for the attachment of other organelle.
- ◆ Site for attachment of ribosomes for photosynthesis.
- ◆ It provides space for glycoprotein & folding of protein.
- ◆ Site for protection and storage of glycogen, steroids and other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

**Endoplasmic Reticulum (ER) Structure**

### 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 prokaryotes 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) dictyosomes are usually found scattered throughout the cytoplasm.
- ◆ **Morphology:** It is a membranous organelle composed of stacks 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 trans-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 channels, 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

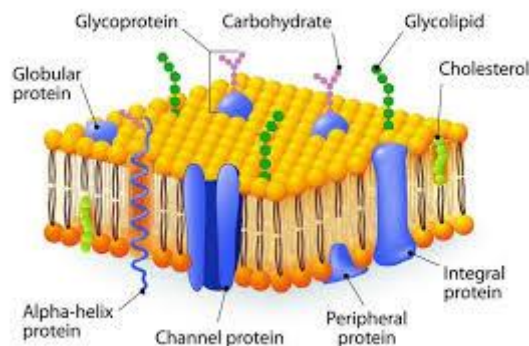
- ✓ Main function is of distribution and shipping for the cells chemical products. Synthesis and secretion of polysaccharides
- ✓ Formation of plant cell wall.

- ✓ 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 reticulum.
- ✓ 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 materials in animals & plant cells.
- ✓ In plants, Golgi apparatus is mainly involved in the secretion of most of primary & secondary cell walls. During cytokinesis of mitosis and 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
  - Solubility properties
  - Ions of different charge
- ☑ Lipid bilayer is about 6.0 nm & 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 fluid 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) Peripheral proteins** - They are bound with electrostatic & H<sub>2</sub> bond Interactions.
  - 2) **Intrinsic (or) Integral proteins** - They internal and bond electrons Hydrophatic interactions.

They belong to 3 categories:

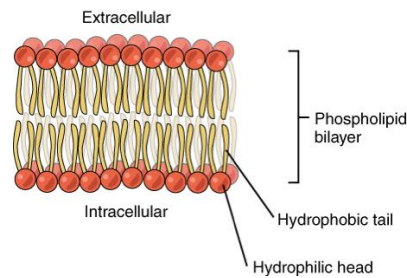
- 1) **Marker Protein**
- 2) **Transport Protein** All float in the lipid layer and move freely
- 3) **Receptor protein**

#### 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 H<sub>2</sub>O soluble molecules and ions.
- ☑ It involves in holding cellular material



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

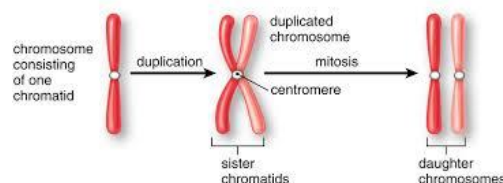


## 1.5 CHROMOSOME

- ☑ Karl Nageli (1842) first observed rod 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 physical 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
  - Cell division
  - Heredity
  - Mutation
  - Repair
  - 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**

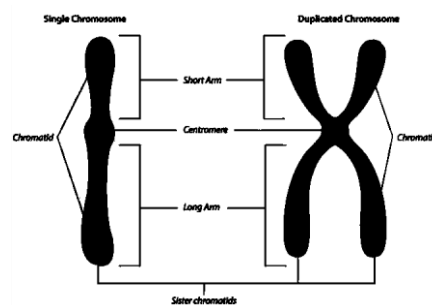
Each cell has pair of each kind of chromosome c/a Homologous chromosome. Chromosomes are made up of Chromatin which contains DNA & Proteins. Each Chromosome contains 100s to 1000 genes. & Lode for many proteins structure of chromosome can be best seen during cell division.



**Main parts of Chromosome:**



- i) **CHROMATIDS:** Each chromosome has two symmetrical structures called Chromatids which is visible in Mitotic Metaphase. But Anaphase 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 Constriction & Nucleolar Organiser's :** This can be identified at Anaphase from centromere. Certain genes in 20 constriction form nucleids. These 20 constrictions are called as Nucleolar 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:** Chromosomes are made up of chromatin i.e. it is made up of DNA, RNA proteins. At Interphase, chromatin fibers are present in nucleoplasm and chromosomes are visible. After staining, darkly condensed region of chromatin is called Heterochromatin 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.





## SPECIALIZED CHROMOSOME

### I. LAMPBRUSH CHROMOSOMES

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 meiosis. These loops give it a brush like appearance. Hence are called lampbrush chromosomes. In certain Urodeles 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.

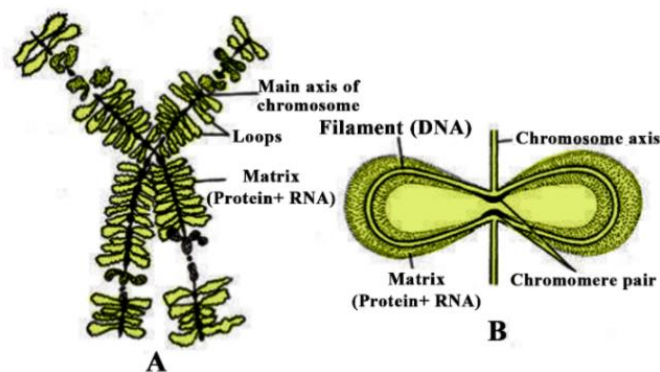


Fig. Lampbrush chromosome. A, Enlarge view of a part of lampbrush chromosome.  
B, One loop of a lampbrush chromosome

- Main axis:** It is the main line containing DNA and proteins and is continuous 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.
- Lateral loops:** Main axis is composed of 4 chromatids (or) 2 bivalent chromosomes. The chromonema of these chromatids gives out lateral globular out growth, called loop axis. Around a loop axis is present matrix made of RNA and proteins which gives it fuzzy appearance. At the base of each loop are darkly stained chromomeres. The diameter of loop axis ranges from 30 to  $50 \frac{0}{A}$  Suggesting two double helices.

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

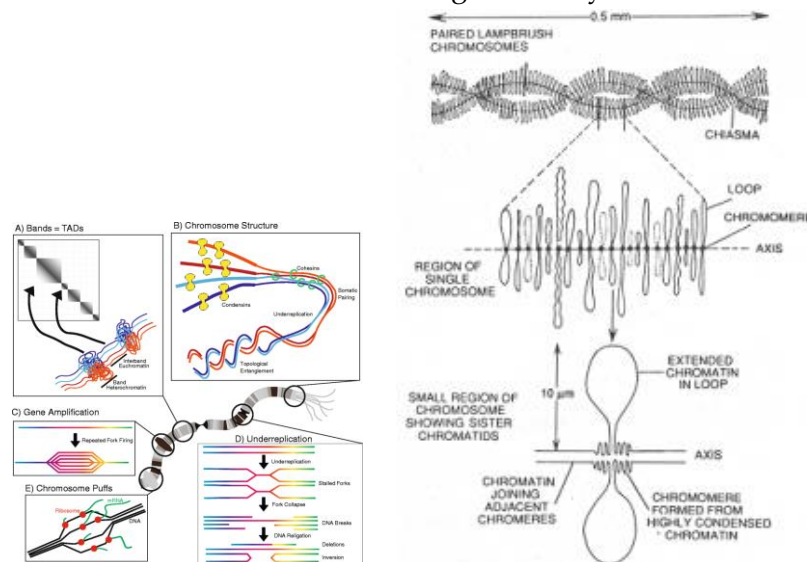
Near the chromomere, matrix is thicker at one end, forming thicker insertion and thinner on the other side forming thinner insertion. Those loops may correspond to genetic loci.

**Functions:**

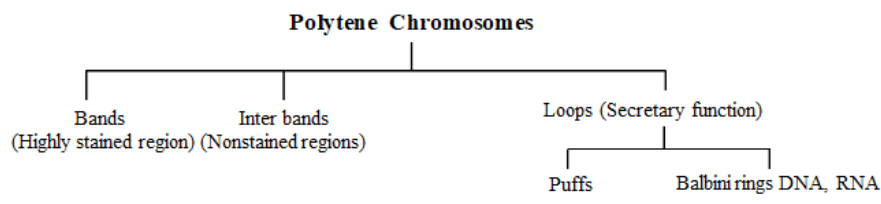
- a) **Synthesis of RNA:** The functions of the lampbrush chromosomes involve synthesis of RNA and protein by their loops. RNA is synthesized only at the thin insertion and then carried around the loops to the thick insertion.
- b) **Formation of Yolk Material:** Lamp brush chromosomes help in the formation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

**II. POLYTENE CHROMOSOME**

Structure of polytene chromosomes: These are relatively smaller than the lampbrush 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 chromomeres of individual chromonemata thus, the bands are due to a tighter 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 swellings or puffs. The metabolic activities, required for the formation of puffs, are related to the secretory function of the salivary glands. This chromosomal RNA differs from the nucleolar and cytoplasmic RNA.

Some regions show large puffs than others, which are called balbiani 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 puffs 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 anaphase.

**Polyploidy:** Extra sets of chromosome but fatal in Humans

**Aneuploidy:** 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 - Monosomy Generally Autosomal Aneuploidy gets aborted

### Aneuploidy 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 develop      Sterile
  - Reduced Mental abilities
3. XYY male (XYY Syndrome)
  - Very tall
  - Heavy acne
  - Aggressiveness
  - Mild mental Retardation      fertile
4. XXX - female (Triple X syndrome)
  - More like XX females
  - Having 2 Barr bodies in sometime cell
  - Mostly sterile, if fertile, rises either XXY or XXX children



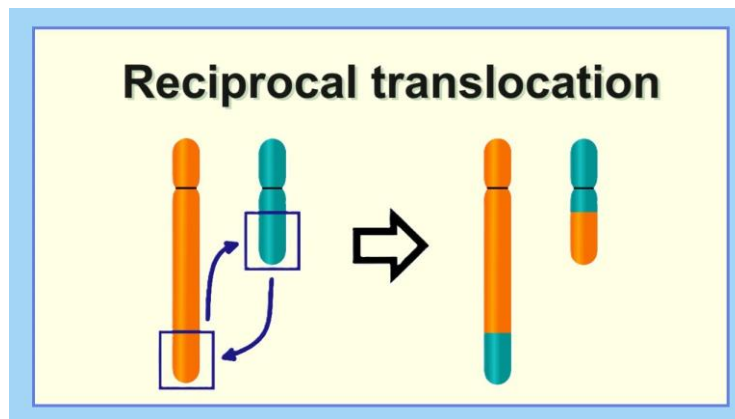
### Aneuploidy in Human Autosomes

1. Autosomic monosomy: Fatal, usually 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
  - ☑ Abnormal facial appearance
  - ☑ Mental retardation
  - ☑ Develop Leukemia & Alzheimer's Disease
  - ☑ Hypotonia - poor muscle tone
  - ☑ Short & broad hands

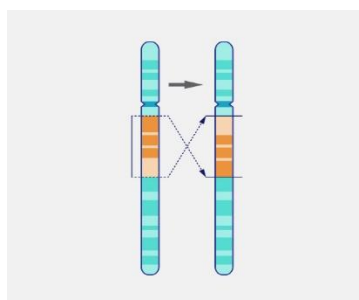
### Abnormalities In Chromosomes And Structure & Rearrangements

#### 1. RECIPROCAL TRANSLOCATION

A portion of 'Chromosome transferred to another.



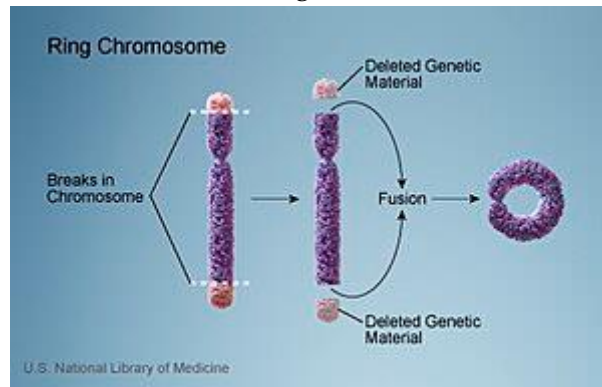
2. **INVERSION:** Inversion of portion of chromosome, broken off turns upside down & reattaches.



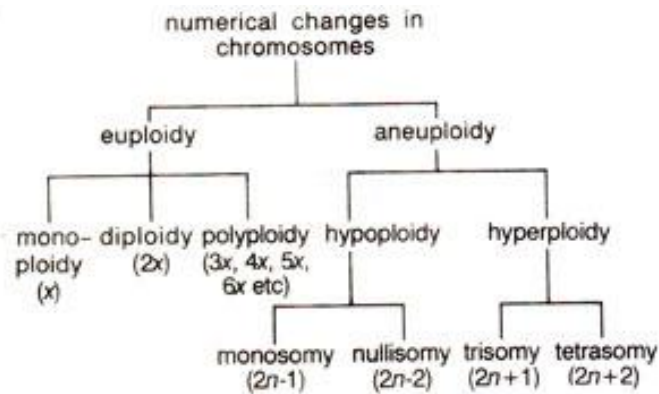
3. **DELETION:** A portion of chromosomes is missing.
  - Eg: i) Wolf Hirschhorn syndrome, deletion in short arm of Ch-4
  - ii) Jacobsen syndrome: Ch-11 'q' arm is deleted in sperm
4. **DUPLICATION:** Portion of chromosome is duplicated.



- 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.

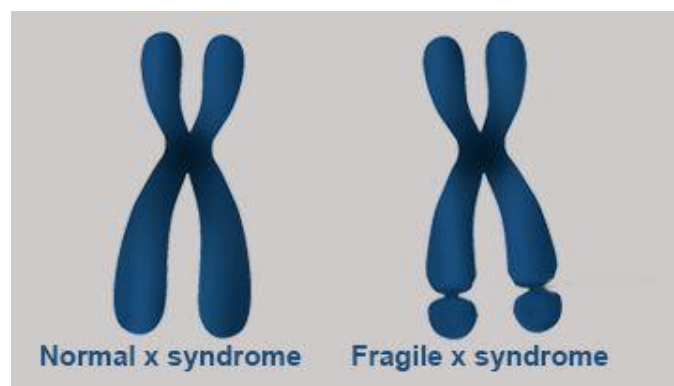


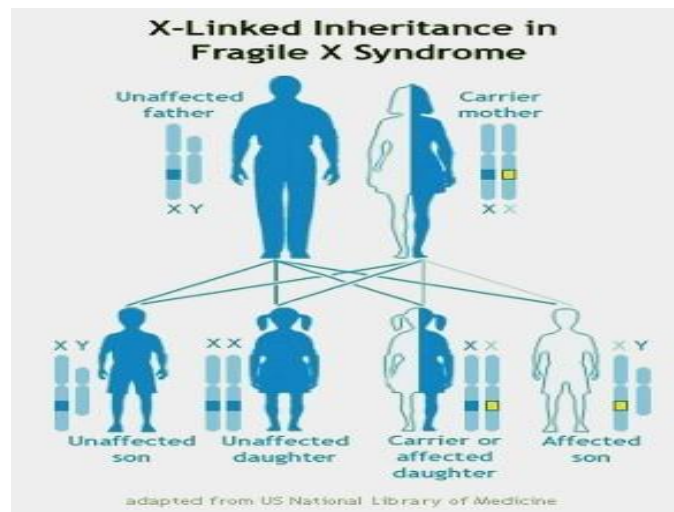
**Numerical changes in Chromosome:**



**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 called as 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.



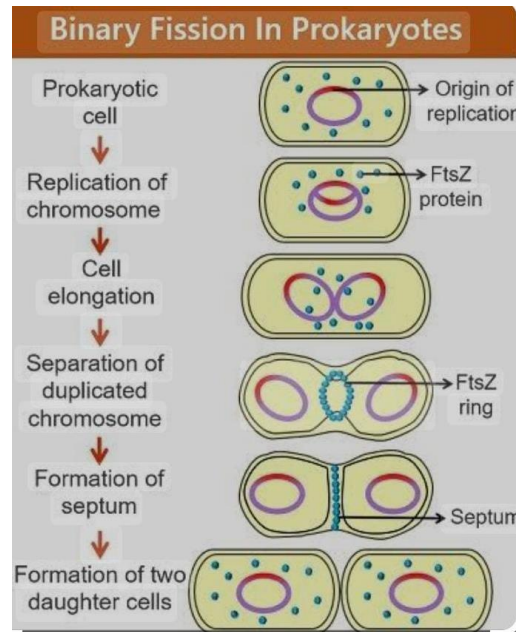
- As replication continues, the 2 origins move at opposite ends of the cell, pulling the rest of the chromosome and thus separation of newly formed chromosomes.

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.

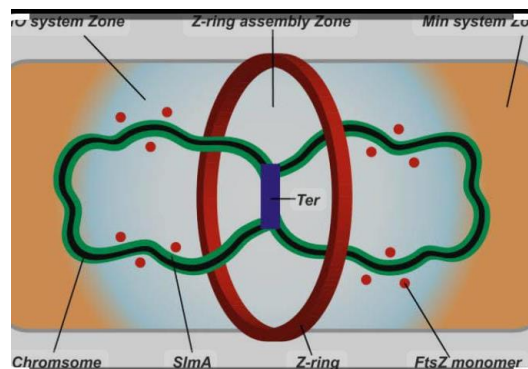
### PROTEINS INVOLVED IN BINARY FISSION

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



**Steps**

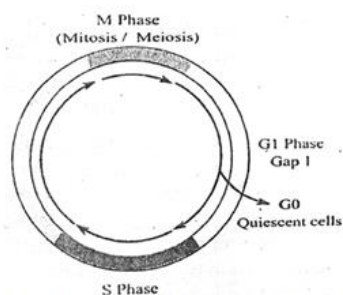
1. DNA is replicated
2. DNA attachment and cell growth
3. Pinching of cell membrane
4. Division of cells
5. Two daughter cells

**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 and then after a brief 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 undergo 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<sub>1</sub> Phase**

- ☑ This is the first stage of interphase, the duration of G<sub>1</sub> phase is highly variable. It may last for minutes (or) week, month (or) year.
- ☑ Metabolic activity is minimum. The enzymes necessary for DNA synthesis during S-phase are synthesized during G<sub>1</sub> - phase cell that never divide again such as Nerve cell are arrested in this stage.
- ☑ Duration of arresting cell at G<sub>1</sub> phase is at specific point. Such state is referred as G<sub>0</sub> state. Cells which pass G - check point can proceed to S-phase.

▲ **'S'-Phase:**

- ☑ It is also called as DNA synthesis.
- ☑ In this 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 initiated at several location ori-site, synthesis of RNA and proteins however expect histones which is required for packing of DNA to form chromatin.
- ☑ The DNA content of Normal cell = 2n but by the end of S-phase the DNA content become = 4n.

▲ **G<sub>2</sub> - phase:**

- ☑ It is also called as post DNA synthesis.
- ☑ This phase is the period between end of S-phase and beginning of prophase. In this period synthesis of RNA & protein occurs for the formation of spindle fibers which are composed of microtubules. The duration varies but shorter than G<sub>1</sub> and S-phase usually last for two hours. After G<sub>2</sub> - phase the cell enter into m-phase & complete all stages such as prophase, metaphase, Anaphase and telophase, after telophase the cell then enters, Interphase via G<sub>1</sub> , S<sub>1</sub> & G<sub>2</sub>.

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

**Mitosis**

- ☑ The term mitosis means thread in Greek Mitosis cell division was first discovered by Flemming in 1879.
- ☑ In Mitosis 2 chromatids of each chromosome separate and move 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 Prophase, Metaphase , Anaphase, 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 and in the metaphase the chromosomes become shorter & thicker, & moves towards nuclear envelope, disintegrates & reduction of size of nuclei. In cytoplasm spindle fibers from centrioles. 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 twisted around each other can be separated when rotated.
- b) **Paraneomic coiling:** They are not twisted around but remain attached. They cannot be separate when rotated.

**Metaphase:**

Complete dissolution of nuclear membrane & arrangement of all chromosomes on the equatorial plate between the two spindles at the poles. Each chromosomes of both poles get attached by spindle fibers each chromatids made up of two sister chromatids. Each sister chromatids connects to spindle fibers 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 chromatids
- ☑ Absence of nucleolus.
- ☑ Disappearance of nuclear membrane.
- ☑ Arrangement of chromosomes on equatorial plate & called as Metakinesis, kinetochores formation by centromere.
- ☑ Condensed state of chromosomes.
- ☑ Spindle fibers centrioles are formed.
- ☑ Centrioles present on both the poles.

**Anaphase:**

In this phase, the chromosomal fibers of spindle contract leading to shortening of spindle fibers 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 fiber through its kinetochore. The sister chromatids migrate towards opposite poles. Based on position of centromere there are 3 types.

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

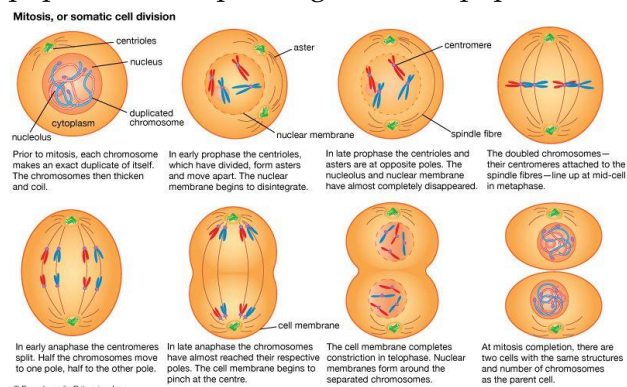
The spindle fibers which pass from one pole to another is called continuous fiber. The spindle fibers that pass from one pole to the centromere of a sister chromatid are called chromosomal fibers.

### Telophase:

- ☑ The last stage of M-phase.
- ☑ The chromosomes decondense like in prophase. Nucleoli reappear.
- ☑ Nuclear envelope forms around each group of chromosomes. Cytokinesis is initiated.

### Cytokinesis:

- ☑ This stage is also called as cell cleavage.
- ☑ The processes of formation of two daughter cells differ in animal and plant cells.
- ☑ In an animal cell, the equatorial contraction originates from outside the cell and moves inward till the midpoint forming microfilaments and a cell plate. This results in two daughter cells. Microfilaments are made up of actin & myosin.
- ☑ In a plant cell, it occurs by formation of a protoplasm made up of microtubules and Golgi vesicles which is made of protein. The tubules and vesicles fuse to form a primary cell wall of a daughter cell.
- ☑ The fraction of dividing cells in a population of cells is called the 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<sub>1</sub> S G<sub>2</sub> 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 G<sub>1</sub>, S, G<sub>2</sub> 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
- Pachytama
- Zygotema
- Diplotema
- Diakenesis

#### Prophase-I

- ☑ This is the first stage of Meiosis
- Leptonema stage:** Lepto = thin, nema = thread .
- ☑ There is increase in nuclear volume



- ☑ The chromosomes condenses & become visible as fine threads.
- ☑ Proteins required for chromosomal condenses 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 throughout 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 and 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) Tetrad stage (or) four strand.
- ☑ Less % of DNA synthesis takes place.

**Diplotema:** (Diplo-separation of into two)

- ☑ In this stage, homologous chromosomes such bivalent gets separated.
- ☑ The separation is in complex become homologous chromosomes 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 fetus development.

### Diakinesis

- ▲ Chiasmata terminilization is completed.
- ▲ 2 homologous chromosomes of each bivalent are attached at both the telomeres only. Chromosomes further condenses & becomes shorter & thickness.
- ▲ Nucleolus and Nuclear envelope disappears.
- ▲ Spindle fibers 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 chiasma then it forms open ring bivalents.
- ▲ Acrocentric & Telocentric forms Rod bivalent.

**METAPHASE-I:**

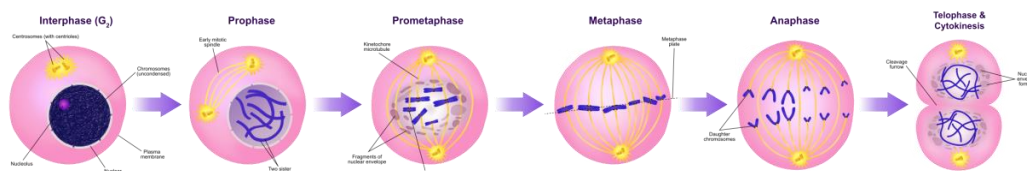
- 1) Nucleolus disappears
- 2) Nuclear membrane disintegration
- 3) Chromosomes arranged in equatorial plate
- 4) Spindle fibers are formed & attached to Kinetochores of sister chromatids.

**ANAPHASE-I:**

- 1) Separation of two homologous chromosomes of each bivalent.
- 2) Migrate towards the poles spindle fibers 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 apparatus are formed two of each of nearly formed.

**Metaphase-II**

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

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

**Telophase-II**

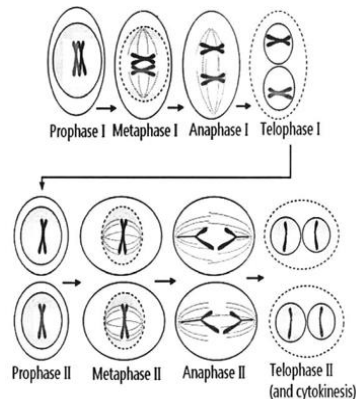
- ▲ Chromatids complete migration and de-condenses
- ▲ Nuclear membrane forms around each of the four nuclear.
- ▲ Each of the four nuclei by the end of telophase-II one parent cell product 4 haploid daughter cells. These 4 daughter cells together is referred as Tetrads.

**Significance of Meiosis:**

- ▲ Production of haploid (n) gametes so that fertilization restores the normal somatic (2n) Chromosomes.

- ▲ Segregations of 2 cells alleles of a gene due to pairing between two chromosomes.
- ▲ Independent segregation of alleles located in separate chromatins at metaphase-I
- ▲ Recombination between linked genes occurs during pachytene stage.
- ▲ Generation of genetic variation through segregation, independent 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 occurring 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 Gametogenesis 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 senescence 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.

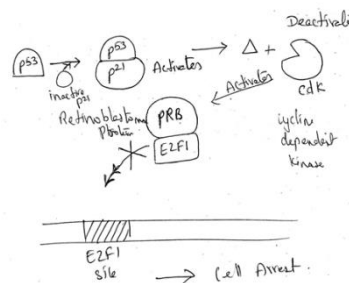
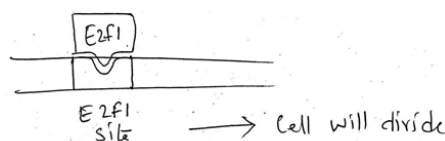
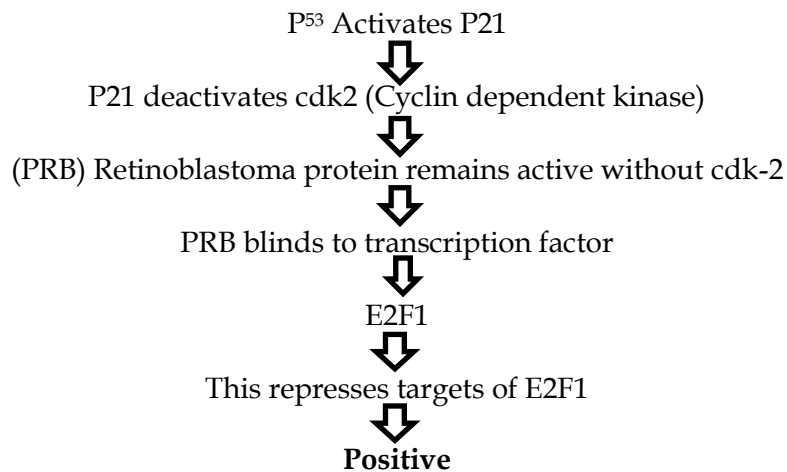
**2.5.1. SENESCENCE**

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 metabolically 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**.

**Signaling Pathways**

1. P<sup>53</sup> pathway
2. P<sup>16</sup> pathway

**P<sup>53</sup> pathway**

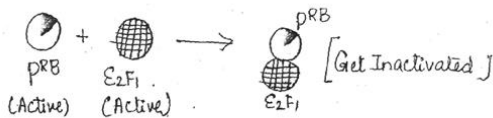
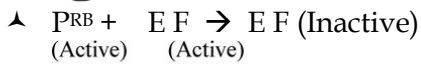
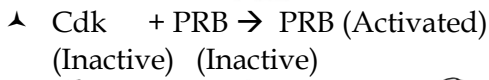
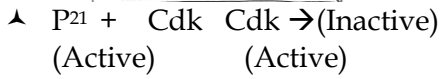
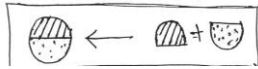
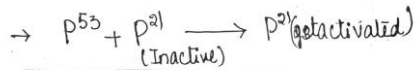


**P<sup>53</sup> Pathway**

- ▲ It is important signalling Pathway in Senescent cells)
  - P<sup>53</sup> - Protein, 53
  - Separated by S.D.S-Page
- ▲ P<sup>53</sup>+ P<sup>21</sup>→P<sup>21</sup> (get activated).



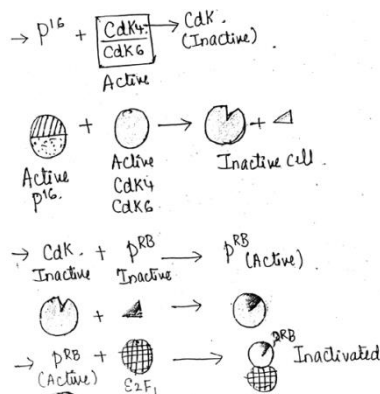
(Inactive)



$P^{53}$  Signalling pathway will make to move normal cell to senescent cell

- ▲  $Cdk \rightarrow$  Cyclin dependent kinen
- $PRB \rightarrow$  Rectino Blastoma protein

**P16 PATHWAY**



P16 is active by inactivating cdk4 & cdk6 by P16  
 (inactive state)  $\rightarrow$  Active PRB binds to E F  $\rightarrow$  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

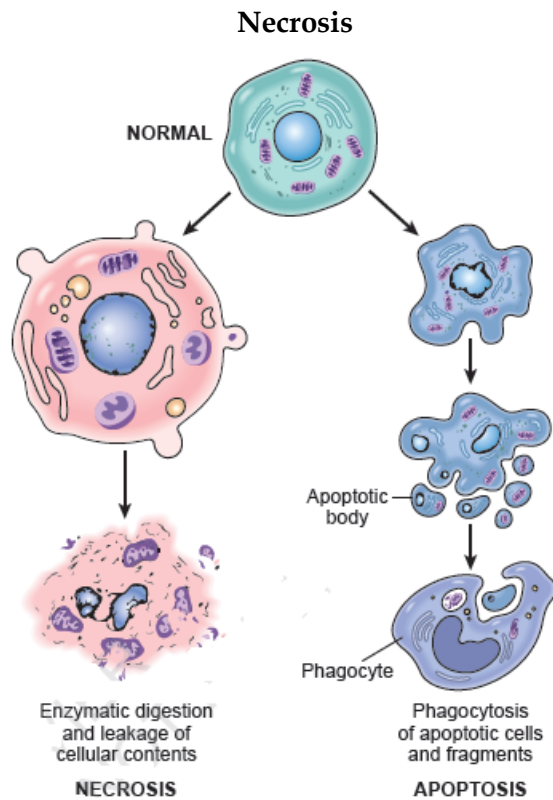
and also by Internal factors such as

- 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 & Liquefactive 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 it causes Pancreatitis.
- 6) **Fibrinoid Necrosis:** Special form of necrosis when antigen binds with antibody which then deposits in arterial walls along with fibrin.

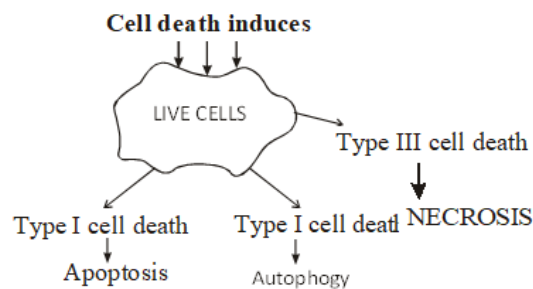




**Treatment for Necrosis**

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**

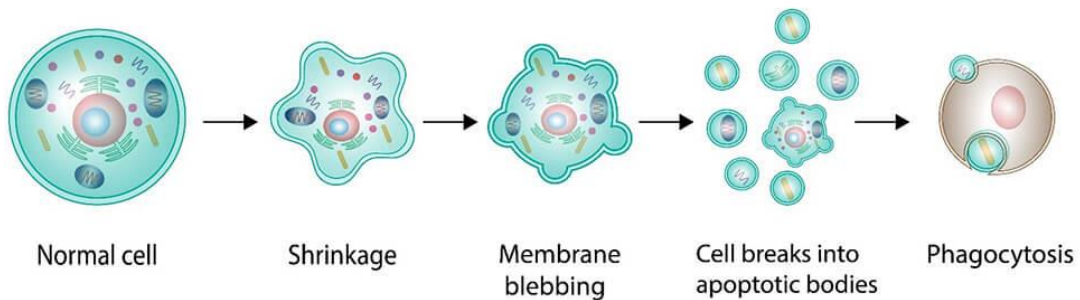
- ◆ Apoptosis is the process of programmed cell death.
- ◆ Biochemical events in the cell during Apoptosis
  - a) Blebbing
  - b) Cell Shrinkage
  - c) Nuclear fragmentation
  - d) DNA fragmentation
- ◆ **In humans (Adult):** 50-70 billion calls die due to Apoptosis in a day 8-14 yrs. of age in children: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

**HISTORY**

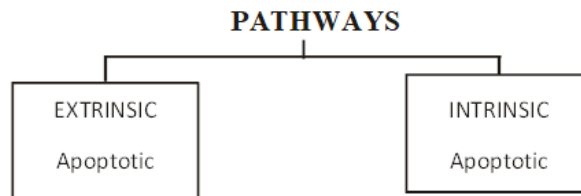
Carl Vogt (1842) German scientist 1st described Apoptosis. The term "Apoptosis" 1st coined by Kerr (1972).

# Apoptosis

- programmed cell death -

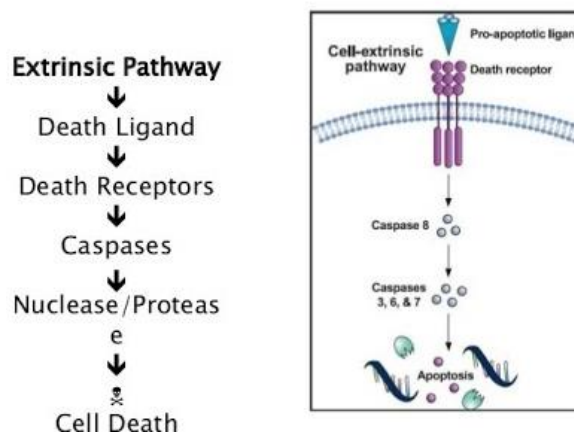


**EXTRINSIC PATHWAY**



- ◆ 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 activates initiator caspases 8, enzyme which are proteases in nature & then activates caspases 3, 6 & 7.
- ◆ Lead to degradation of cellular function & finally causes Apoptosis.

## Extrinsic Pathway

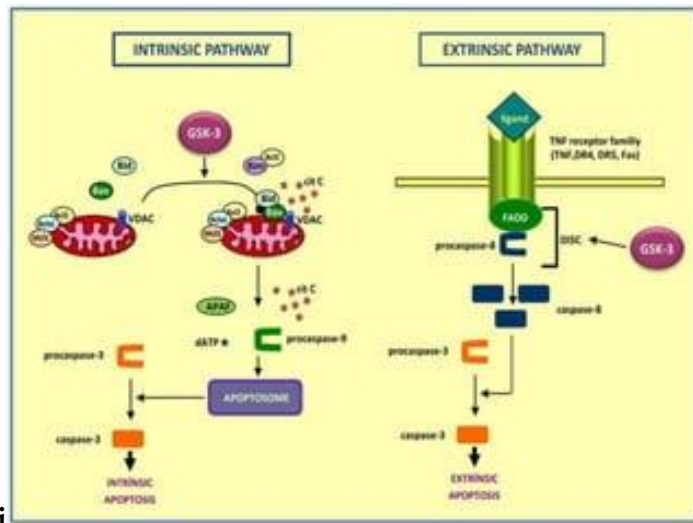


**INTRINSIC PATHWAY**

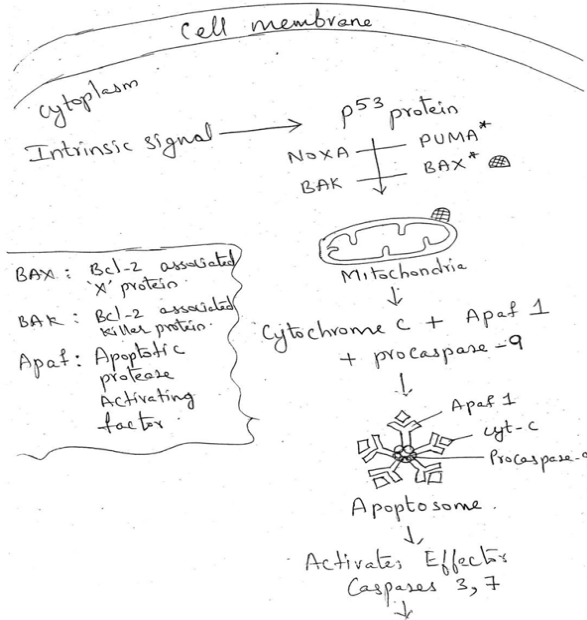
The cell in intrinsic pathway produces P53 protein, where NOXA produces PUMA & BAK converts into BAX.



BAX binds to Mitochondria  
 ↓  
 Produces now Cyt C + Apaf1 + Procaspase 9  
 ↓  
 Apoptosome formation  
 ↓  
 Activates Effector caspases 3,7  
 ↓  
 Leads to blebbing, nuclear fragmentation & Apoptosis  
**INTRINSIC PATHWAY**



ii



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 Sativum*) plants.















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

1. Peas have many distinct, well defined and observable morphological characteristics. (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 within 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 color** - green (or) yellow
  - ◆ **Pod shape** - Inflated (or) constricted
  - ◆ **Stem length** - Tall (or) short
  - ◆ **Seed shape** - Round (or) wrinkled
  - ◆ **Seed color** - Yellow (or) Green
  - ◆ **Flower color** - 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 Characters in Pea Plant Studied By Mendel :**

<b>F<sub>1</sub> Crosses for Seven Characters in Pea Plants</b>					
Character	Dominant Trait	×	Recessive Trait	F <sub>2</sub> 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

*F<sub>1</sub> crosses for seven characters in pea plants and their results in F<sub>2</sub> generations*

The results of Mendel's experiments are

1. The inheritance of each trait of an individual is determined by GENES.
2. For morphological characteristics (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 MENDEL'S PRINCIPLES (OR) MENDEL'S LAW.

1. Law of segregation - Mendel's 1st law
2. Law of Independent assortment - Mendel's 2nd law.

#### LAW OF SEGGREGATION:

For any particular 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

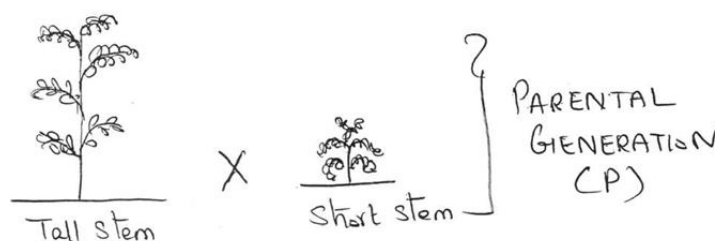
The inheritance of one trait does not affect the inheritance of another.

#### 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 - MONO HYBRID CROSS / RATIO

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

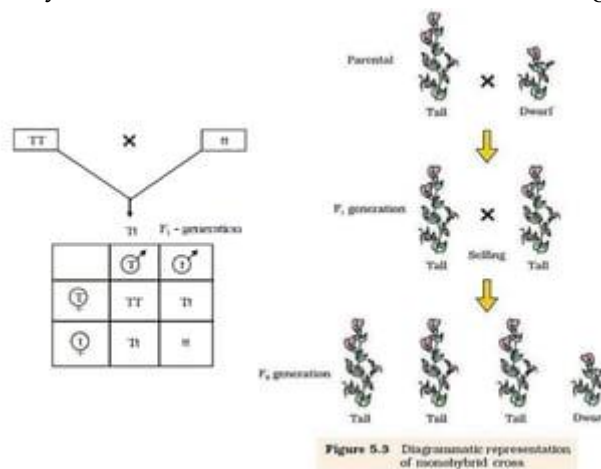


Two different varieties of plants are considered as parental generation. (P). These two plants are artificially cross pollinated to produce first filial generation also called



as F<sub>1</sub> generation. The plants of F<sub>1</sub> generation were sown in soil and allowed the next generation called second filial generation or F<sub>2</sub> 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. Parental generation comprises of tall characters and short character.
4. All plants in F<sub>1</sub> generation are Tall.
5. It appeared that short character was not present in plants.
6. Short character reappeared in some plants in F<sub>2</sub> generation.
7. Statistical analysis showed 75% tall and 25% short. in F<sub>2</sub> generation



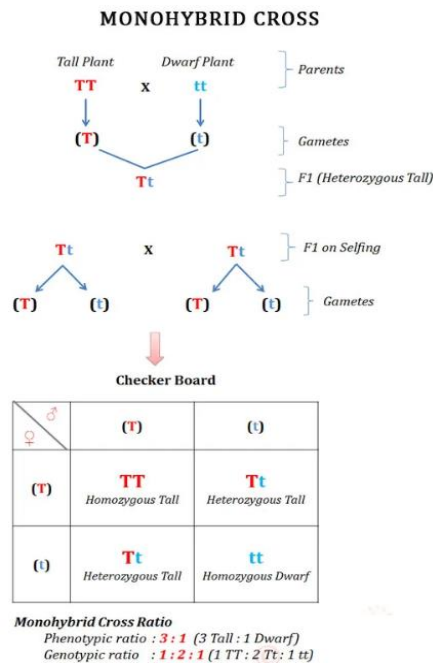
### ANALYSIS OF MONOHYBRID CROSS

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

Example: Tall (TT) : Short (tt)

The contrasting characters for each trait are called Allelo morphs (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 allele '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 INDEPENDENT ASSORTMENT**

DIHYBRID AND TRIHYBRID CROSS inheritance of one trait does not affect the inheritance of another. Breeding experiments involving study of two pairs of contrasting characters are called DIHYBRID CROSS. A cross involving three pairs of contrasting characters are called TRIHYBRID CROSS.

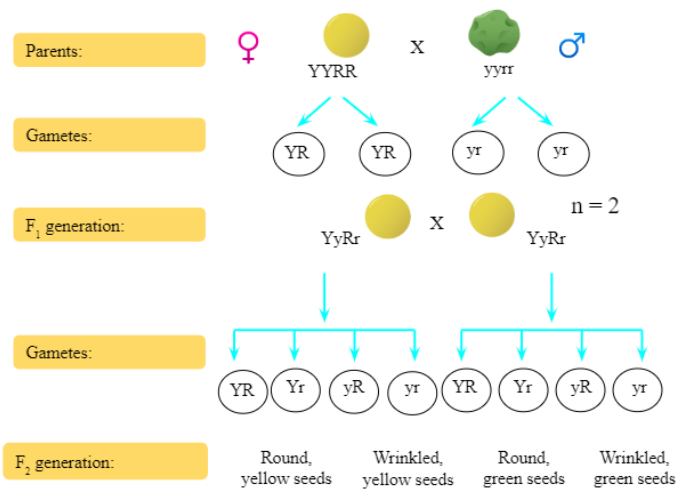
**DIHYBRID CROSS**

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 F<sub>2</sub> generation, four types of combinations were observed.

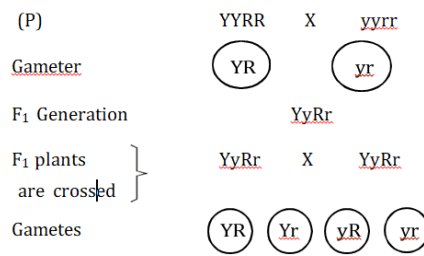
- a) Both dominant trait in 9/16 of the population.
- b) First trait dominant and second trait recessive in 3/16 of the population.
- c) First trait recessive and second trait dominant in 3/16 of the population.
- d) Both traits recessive in 1/16 of the populates

**DIHYBRID CROSS**

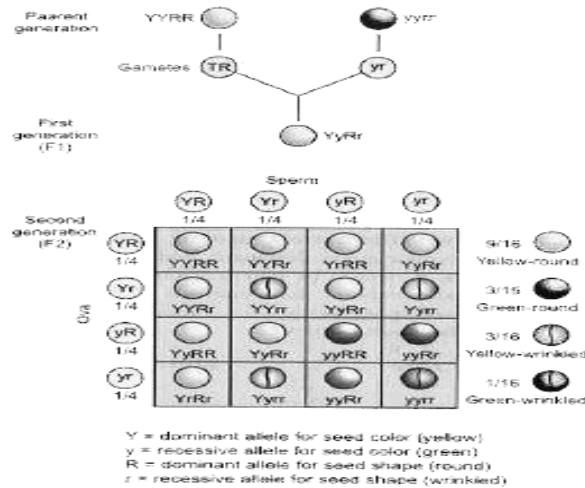


Type	Ratio	Combination
Yellow, Round	9/16	Parental
Yellow, Wrinkled	3/16	Recombinant
Green, Round	3/16	<u>Recombinant</u>
Green Wrinkled	1/16	Parental

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



**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 Independent Assortment states that, when dihybrid form gametes.

1. Each gametes receive one allele from each allelic pair.
2. The alleles are independently 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 characters are called trihybrid cross Trihybrid cross between tall plants bearing yellow and round seeds and short plants bearing green and wrinkled seeds.

Genotype for tall plants with yellow & round seeds - TTY<sup>YY</sup>RR

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

**TRIHYBRID CROSS**

[P] Parents TTY<sup>YY</sup>RR 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	TTYRRR	TTYRRr	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
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**

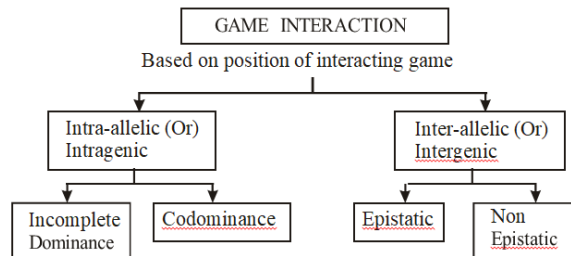
- 27: Dominant phenotype for 3 traits  
 Tall: Yellow: round
- 9: Dominant phenotype for 2 traits  
 Tall: yellow: wrinkled
- 9: Dominant phenotype for 2 traits  
 Tall: green : round
- 9: Dominant phenotype for 2 traits  
 Short : yellow : round
- 3: Dominant phenotype for 1 trait  
 Tall: green : wrinkled
- 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 not necessary that single gene determines a single character, but same character may be affected by more than one gene by Interaction. Such Mechanism of genes are call "GENE INTERACTION".

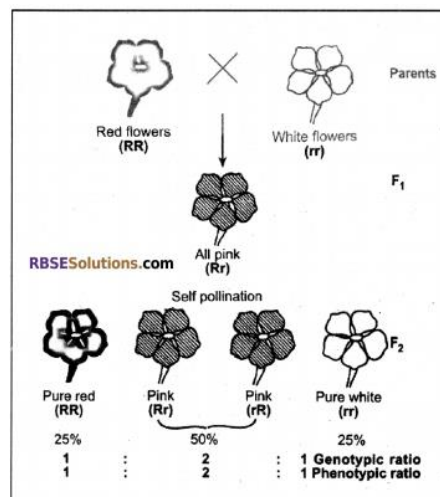
- ▲ **Intragenic** : Interaction of genes is between alleles of same locus.
- ▲ **Intergenic** : Interaction of genes is between alleles of different locus.



### 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:

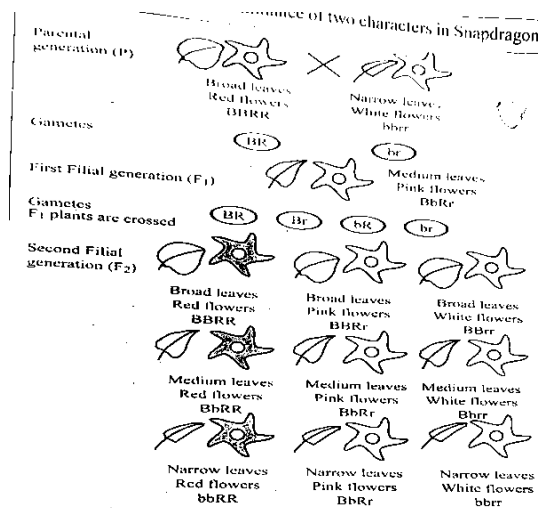


Incomplete dominance in *Mirabilis jalapa*

#### 1. Incomplete dominance of one character of *Mirabilis Jalapa*

In F<sub>2</sub> generation, 25% plants gave red flowers, 25% gave white color flower and 50% plants have pink flowers. Thus phenotypic ratio is modified for Mendel's ratio from 3:1 to 1:2:1. Because allele is dominant over the other.

#### 2. Incomplete dominance of two characters in *Mirabilis Jalapa*.



Punnett Square

	BR	Br	bR	br
BR	BBRR Broad 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 Medium Pink	bbRR Narrow Red	bbRr Narrow Pink
br	BbRr Medium Pink	Bbrr Medium White	bbRr Narrow Pink	bbrr Narrow White

Modified Di hybrid 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 color of Mirabilis plants. A cross between plants with broad leaves and red color flower and plants with narrow leaves and white color flower gives plants with medium leaves and pink flowers in F1 generation.

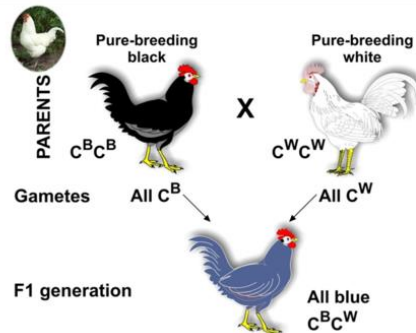
Codominance

If the heterozygotes exhibits mixture 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

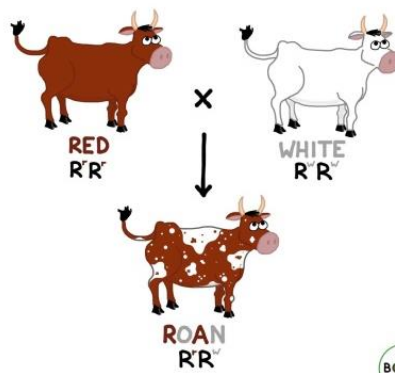
#### Incomplete Dominance – Andalusian Chickens (2)



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

### 2. Codominance in Cattle Coat Colour

## CODOMINANCE



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

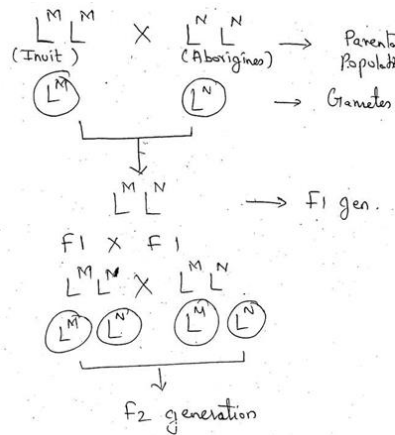
#### 3.3.2 CODOMINANCE

- ◆ Codominance is found in human genes. One best example in MN blood group. MN genes are under the control of Autosomal locus. MN genes express 'M' and 'N' ag. These are under the classification of Glycophorins.
- ◆ MN antigens bind 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 ( $L^N L^N$ )





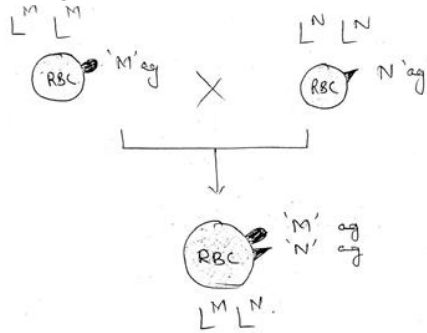
Crossing of Inuit & Aborigines :



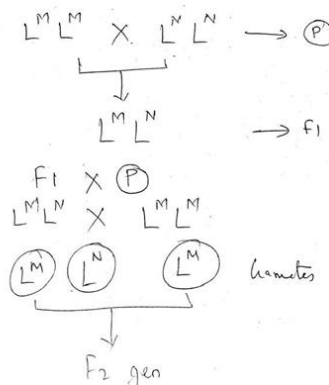
Phenotypic ratio 1: 2 :1

	$L^M$	$L^N$
$L^M$	$L^M L^M$ Dominant Homozygotes RBC - 'M' Ag	$L^M L^N$ Heterozygotes (RV - 'M' 'N')
$L^N$	$L^M L^N$ Heterozygotes RBC $\begin{matrix} M \\ N \end{matrix}$	Dominant Homozygotes (RBC) $^N$

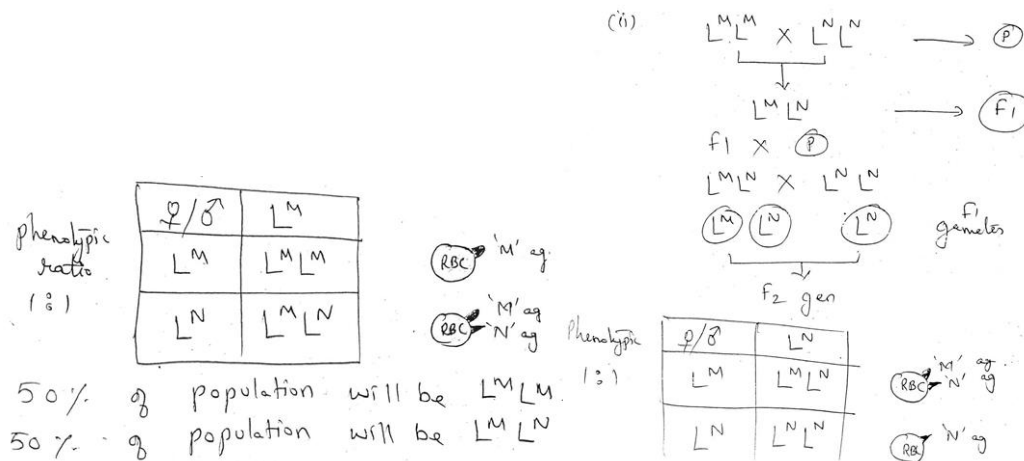
Punnett square for MN blood gp



TEST CROSS



**Punnettee 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 Independent Assortment states that alleles of two different genes are inherited independently resulting in characteristic 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 color 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 fur.

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

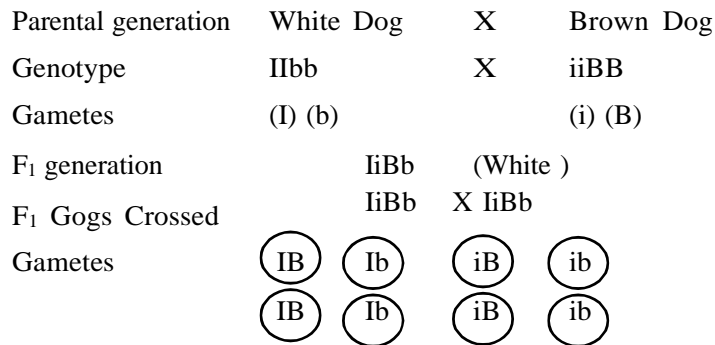
Dominant 'I' = inhibits pigment formation in the fur of dog.

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

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

Recessive 'b' = Produce Black color fur in the dog.

**DOMINANT EPISTASIS SHOWN IN COAT COLOUR IN DOGS**



**Punnett Square**

	I <b>B</b>	I <b>b</b>	i <b>B</b>	i <b>b</b>
I <b>B</b>	I <b>BBB</b> DOMINANT MASKED	I <b>Bbb</b> DOMINANT MASKED	i <b>BBB</b> DOMINANT MASKED	i <b>Bbb</b> DOMINANT MASKED
I <b>b</b>	I <b>Bbb</b> DOMINANT MASKED	I <b>bbb</b> DOMINANT MASKED	i <b>Bbb</b> DOMINANT MASKED	i <b>bbb</b> DOMINANT MASKED
i <b>B</b>	i <b>BBB</b> DOMINANT MASKED	i <b>Bbb</b> DOMINANT MASKED	i <b>iBB</b> RECESSIVE DOMINANT	i <b>iBb</b> RECESSIVE DOMINANT
i <b>b</b>	i <b>Bbb</b> DOMINANT MASKED	i <b>bbb</b> DOMINANT MASKED	i <b>ibb</b> RECESSIVE DOMINANT	i <b>ibb</b> RECESSIVE RECESSIVE

**DOMINANT MASKED : 12** (White color fur)

**RECESSIVE DOMINANT : 3** (Brown color fur)

**RECESSIVE RECESSIVE : 1** (Black color 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	I <b>bb</b>	X	i <b>BB</b>
Phenotype	White		Brown
Gametes	I <b>b</b>		i <b>B</b>
F <sub>1</sub> genotype		I <b>ibb</b>	

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 color in Mice.

1. Coat color is determined by two genes
  - a) Agouti - Agouti gene A
  - b) Black - Black gene C
2. Three different colors are formed by the combination of these two genes.
3. Agouti color 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 characteristic banded pattern. Hairs are black with yellow bands near the tip of the hair.
4. Black color 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 color 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 color.

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 pigment, 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 (Recessive masked)

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

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

**3. DUPLICATE DOMINATE EPISTASIS (15: 1)**

This made of epistasis are also called "duplicate action". The presence of dominant allele, 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 Shepherd's purse plant

1. There are two types of seed shape in Shepherd's purse plant.
  - a) Triangular
  - b) Elongated or top shaped
2. The genes controlling these characters 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 elongated 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-
Phenotype	Triangular	Triangular	Triangular
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.

#### 4. DUPLICATE RECESSIVE EPISTASIS: (9 : 7)

This is also known as complementation 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 independent gene. Thus modified dihybrid ratio are formed.

**Example:** Flower Color of Sweet Pea : Dominant color of sweet pea flower are purple. This color is dependent on two non-allelic complimentary genes. They are

- 1) Gene C 2) Gene P.

The product of gene 'C' is a color less substance. The product of gene 'P' transforms colorless substance to purple color. This means both gene 'C' and gene 'P' are required for purple color 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	$3/16+3/16+1/16=7/16$		

$\frac{\text{♀}}{\text{♂}}$	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.

Inhibition of color formation (due to presence of dominant I) and absence of color (due to absence of dominant allele at 'C'), both produces white plumage as



phenotype. To produce colored plumage, the genotype at first locus should be  $ii$  and second locus should be 'C'.

### 5. 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 color swine

AA → Produces sandy coat color

BB → Produces sandy coat color

AB → Produces Brown coat color

ab → Produces No color 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 influenced 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 color flower from number of plants, all flowers have same shade of violet. There will not be deep violet or pale violet flowers.

#### Variable Expressivity:

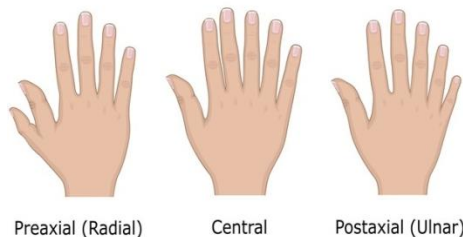
- ◆ Variation in expressivity is due to the presence of complementary, supplementary or modifier (or) suppressor gene.
- ◆ The gene for polydactyly, results in production of more than 5 fingers or toes per hand or foot & has 70% penetrance.

Human Polydactyly: {

- Postaxial (little finger)
- Preaxial (Thumb)
- Central

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

#### Types of Polydactyly



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) of soft tissue

#### Syndromes associated:

- Down syndrome, syndactyly (joint fingers) etc.

#### WARDENBERG SYNDROME

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

#### Type 1 : (WS-1) posses

- Congenital hearing loss
- Forelock of white hair. (Poliosis)
- Different colored eye (both eyes)
- Heterochromic iridum





- Different color eye (multiple eye)
- Sectoral heterochromia iridum
- Patches in skin depigmentation
- Wider gap between inner corners of eyes - Telecanthus.
- High Nasal bridge.
- Flat nose tip
- Smaller edges of the nostrils.

**Type-2 (WS-2)**

- Smaller eyes (microphthalmia)
- Hardened bones (compact bones)
- Deafness.
- Under development of inner - ear structure.
- Anosmia (lack of sense of smell) due to back of olfactory bulb in the brain.

**Type - 3 (WS-3) (or) Klein - Waardenburg 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 - Waardenburg syndrome )**

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

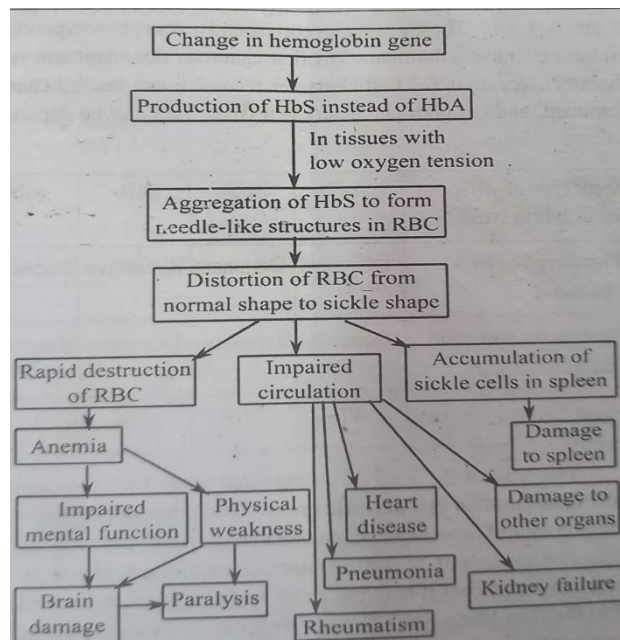
The phenomenon of single gene influencing more than one character is called pleiotropic and such gene are called pleiotropic genes.

Examples for pleiotropism are

1. Sickle cell anemia
2. Phenylketonuria
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 Haemoglobin 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 Schmidt 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) Malnutrition
  - 6) Exposure to drugs, alcohol.

## Signs & Symptoms

- ▲ Head circumference is smallest than normal. Dwarfism.
- ▲ Short stature Facial distortions
- ▲ Balance & coordination problem Hyper activity
- ▲ Neurological problem, Zika viral Infection
- ▲ Treatment
- ▲ 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 infections

**Risk Factors**

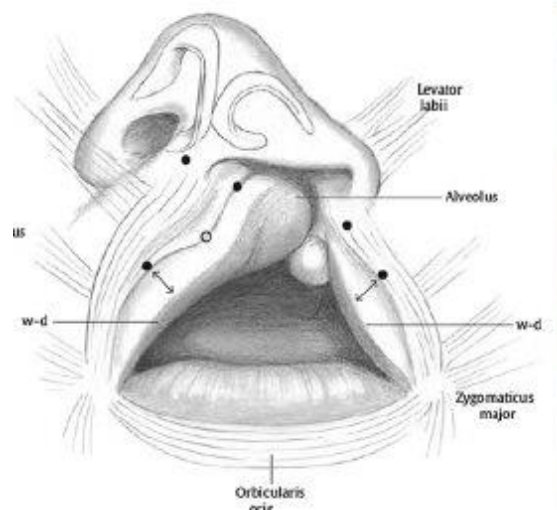
- 1) Family history
- 2) Exposure to certain substances during ncy.
- 3) Having Diabetes
- 4) Being obese during Pregnancy

**Complications:**

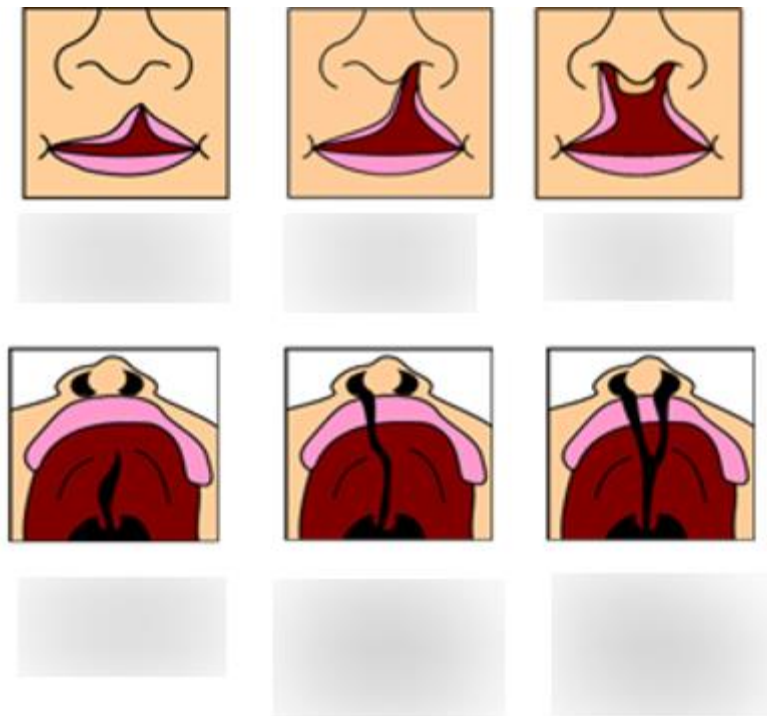
- ▲ Difficultly in feeding
- ▲ Ear infections
- ▲ Hearing loss
- ▲ Dental problems
- ▲ Speech difficulties

**Preventions :**

- ▲ Consider genetic counseling
- ▲ Take prenatal vitamins
- ▲ Do not use tobacco (or) alcohol

**CLEFT LIP**

Types of Cleft Lips



3.5 MULTIPLE ALLELEISM

An allele of a genetic locus having more than two allelic forms with in a 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:

- Multiple alleles in Rabbits for coat color 4 alleles exist in 'C' gene.  
 wild - type version - C<sup>+</sup> C<sup>+</sup> expressed as Brown - for chinchilla phenotype - C<sup>ch</sup> C<sup>ch</sup> expressed as Black tipped white fur  
**Himalayan phenotype C<sup>h</sup> C<sup>h</sup> Black fue on extremities & white for Albino, recessive phenotype - cc expressed with white fur.**

Albino can also be expressed as C<sup>a</sup> C<sup>a</sup> .

**Coat color of Rabbit:** It is inherited as a series of multiple alleles. In case of coat color of Rabbits, there are 4 alleles and each one is expressed with a different phenotype.

The order of Dominance is C > C<sup>ch</sup> > C<sup>h</sup> > C<sup>a</sup>

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

C<sup>h</sup> C<sup>a</sup> X C<sup>h</sup> C<sup>a</sup>----- (p)  
 (C<sup>h</sup>)(C<sup>a</sup>) (C<sup>h</sup>)(C<sup>a</sup>)----- Gemetes

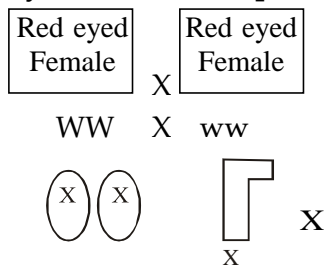
1<sup>st</sup> Cross

$C^h C^a \rightarrow F_1$   
 $F_1 \times F_1$   
 $C^h C^a \quad C^h C^a$

	$C^h$	$C^a$
$C^h$	$C^h C^b$ (H)	$C^h C^a$ (H)
$C^a$	$C^h C^a$ (H)	$C^a C^a$

H-Himalayan  
 A-Albino

Eye color of Drosophila



1<sup>st</sup> cross

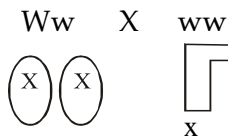
$F_1$

	W	W
w	$Ww$	$Ww$
w	$Ww$	$Ww$

$\rightarrow$  Red Eye Female  
 $\rightarrow$  Red



Red eyed female  $\times$  red eyed male



		$X$ $W$	$X$ $w$
$X$	$W$	$WW$  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.
- Dominant eye color is Red recessive eye color is white
- But Heterozygotes produces other shades from white to red, wine, blood, eosin, honey, pearl, lorry.
- In F2 generation, Mutant strains show variation in wide range color between

red to white.

**ABO Blood Group:**

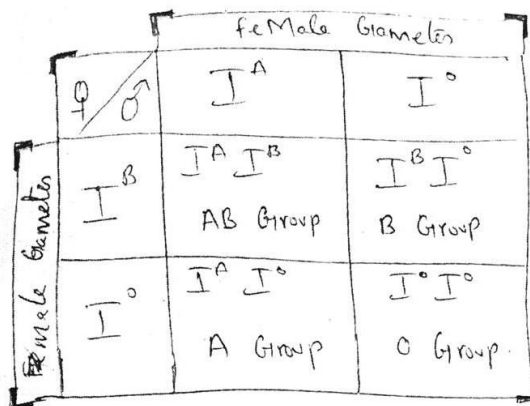
Blood groups are inherited from both parent. There are three alleles for ABO blood groups in humans. If chromosome contains allele A, then protein A is produced, then the RBC's of that individual will contain protein A on its surface. And that individual falls under 'A' Blood group likewise for allele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

**Types of blood groups & its Genotype**

A	$I^A I^A$ $I^A I^0$	(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.

$$I_A I_0 \times I_B I_0 \text{----- (P)}$$



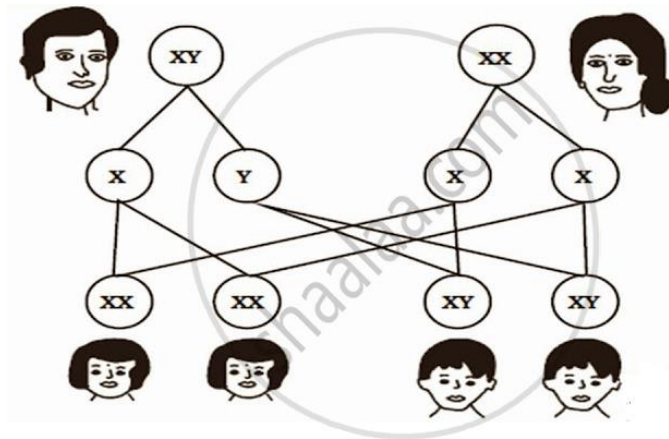
phenotype ratio  
Genotypic ratio  
1:1:1:1

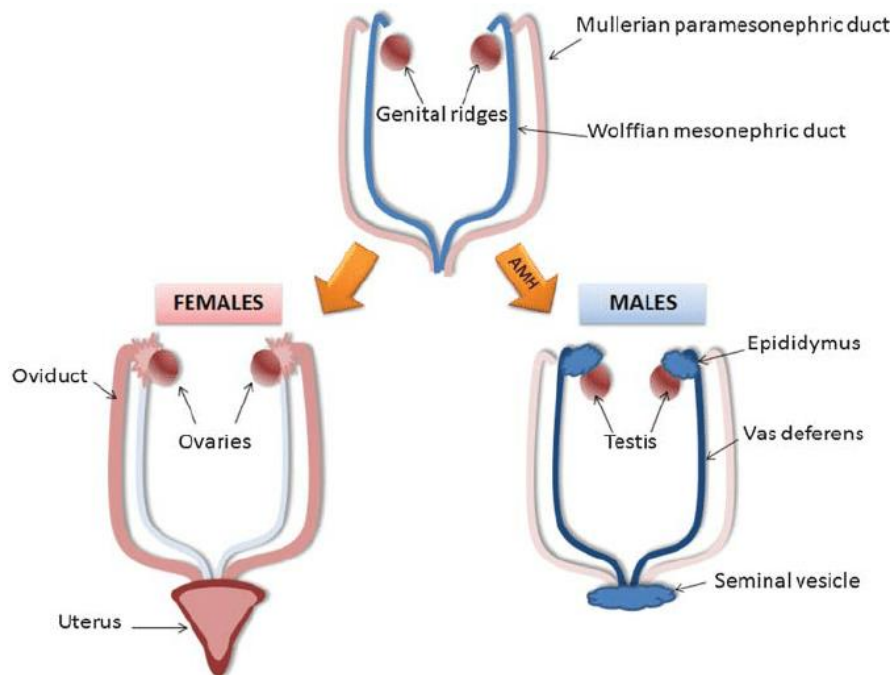
**Example:** When Homozygote of O group (♀) cross with group AB. Find out the possible blood groups in the off spring.

$I^o I^o \times I^A I^B$

		Female		
	♀	$I^o$	$I^o$	Phenotypic ratio 2 : 2. Genotypic ratio 1 : 1 : 1 : 1
	♂	$I^A I^o$ A Group	$I^A I^o$ A Group	
Male.	$I^A$	$I^A I^o$ A Group	$I^A I^o$ A Group	
	$I^B$	$I^B I^o$ B Group	$I^B I^o$ B Group	

### 3.6 XY Chromosome

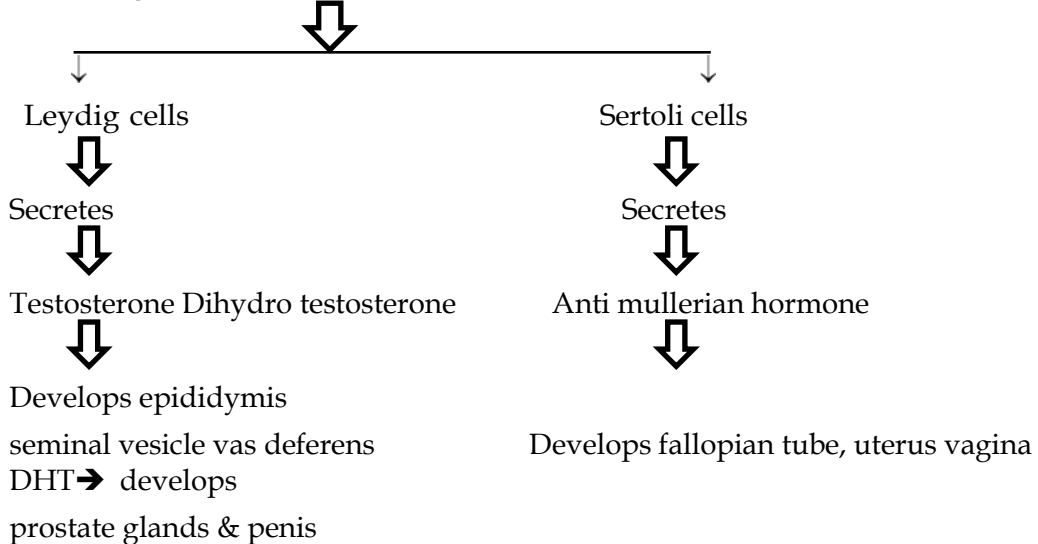




Sex-determining region of Y chromosome in embryonic germ cells (SRY gene)

↓  
SRY protein - i.e. Testis determining protein

↓  
This causes gonad medulla to differentiate into testis



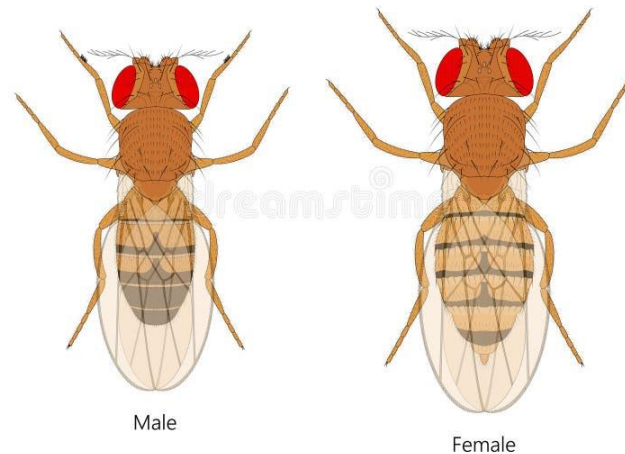
**Sex Determination In Drosophila**

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 protein. While in producing female drosophila, complete removal of I<sub>1</sub> & I<sub>2</sub> makes the mRNA



to produce SXL protein. This protein activates 'Tra gene and produce tra protein. Later Tra protein regulator DSX gene to produce female double sex proteins. This mRNA regulation to produce female double sex protein.

### *Drosophila melanogaster*



#### **3.6.2 Sex Determination in Human**

- ▲ Sexual differentiation in humans is the process of development of sex differences 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.

<b>Female</b>	Contains	XX	Chromosome
<b>Male</b>	Contains	XY	Chromosome

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

**Female** - Para Mesonephric ducts

**Male** - Mesonephric ducts

Until seven weeks after fertilization, the fetus appears to be sexually indifferent, i.e. the fetus neither looks like male or female, but over next 5 weeks, the fetus begins to produce Hormones that causes sex organs. 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.



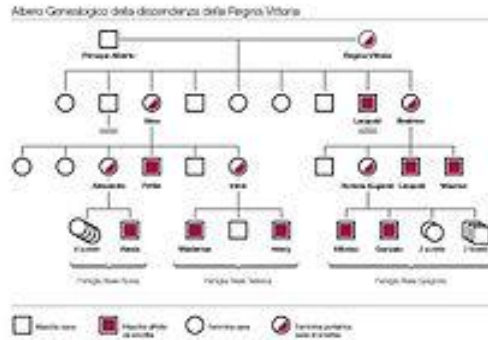
**3.6.3 X- Linked Inheritance**

**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.

- Hemophilia A** (Classic hemophilia or clotting factor gene factor VIII deficiency) is inactivated
- 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.

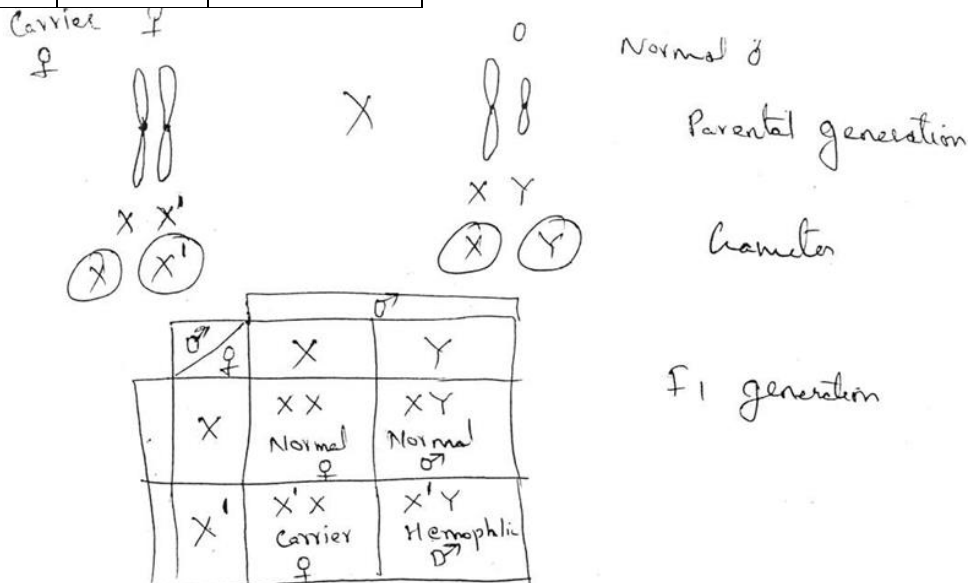


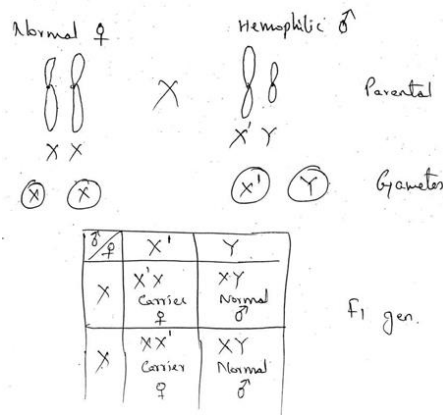
When assigning alleles for sex linked traits for hemophilia.

$X^H$  - Dominant (X)

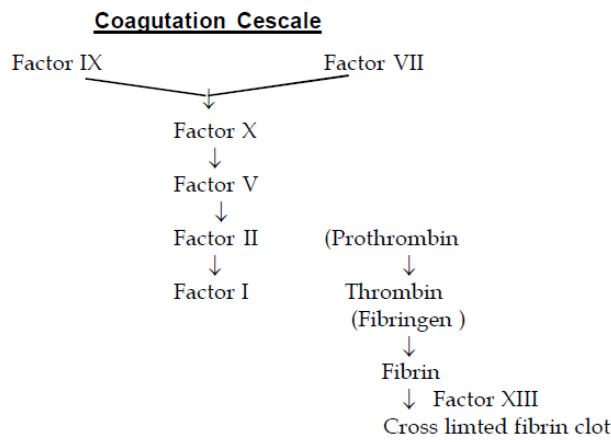
$X^h$  - recessive ( $X^1$ )

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

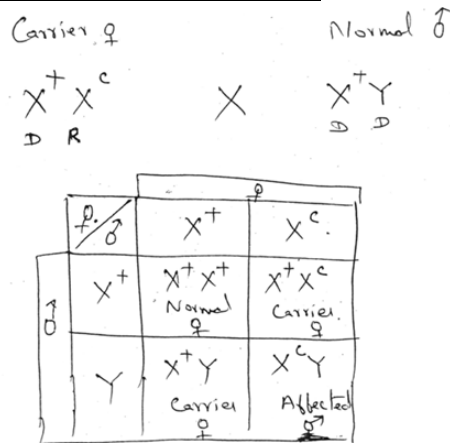




**Coagulation Cascade**



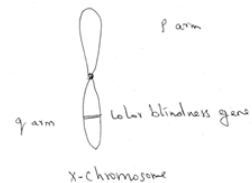
**3.6.4 Color Blindness (Red - Green Color blind)**



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 normal.

Males are affected more than females.

- ▲ X<sup>+</sup> or X<sup>N</sup> is dominant
- ▲ X<sup>c</sup> or X<sup>n</sup> is recessive



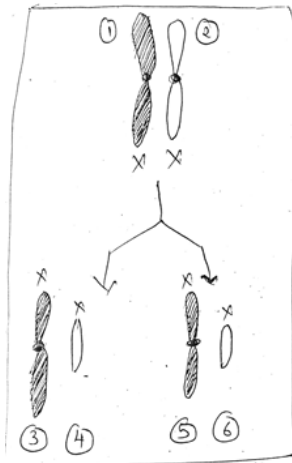
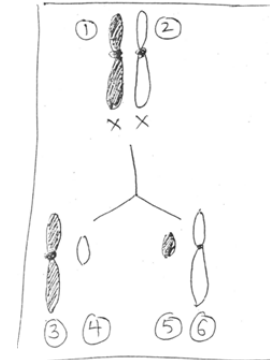
**3.6.5 X - Inactivation or XCI (X- Chromosome inactivation)**

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).

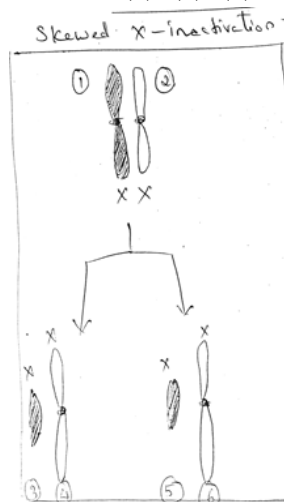
1. Maternal X chromosome
2. Paternal X chromosome
3. Maternal X chromosome is normal while
4. Paternal X chromosome is inactivated
5. Inactivated Maternal X chromosome
6. Normal paternal X chromosome

In both the offspring, 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 - chromosome inactivate
- Paternal X - chromosome is normal (1), (4), (6)



**X-inactivation Example For Random - Calico Cat**

Heterozygous : Black & Orange

CAT (X<sup>B</sup>) (X<sup>0</sup>)

X<sup>0</sup> X<sup>B</sup>

X<sup>b</sup> X<sup>b</sup> → Black females cat

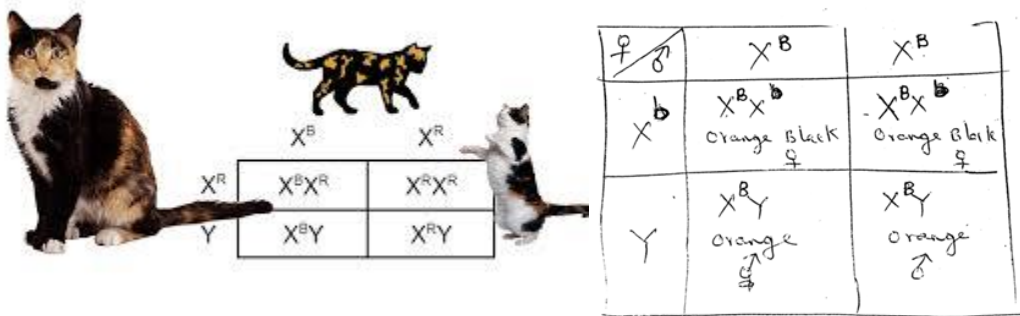
X<sup>B</sup> X<sup>B</sup> → Orange females : cat

X<sup>B</sup> Y → Orange males cat

X<sup>b</sup> Y → Black male cat

X<sup>B</sup> X<sup>b</sup> → Black & orange cat female (calico cat)

X<sup>B</sup> X<sup>B</sup> (orange cat)      X<sup>b</sup> Y (Black cat)



**Mechanism**

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



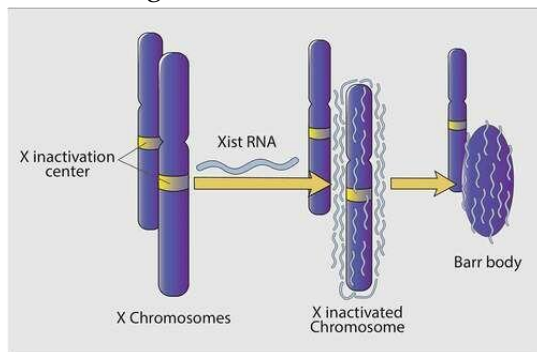
This process is controlled by

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. Random 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 coupling and repulsion are the aspects of same phenomenon called as LINKAGE.

### COUPLING AND REPULSION THEORY

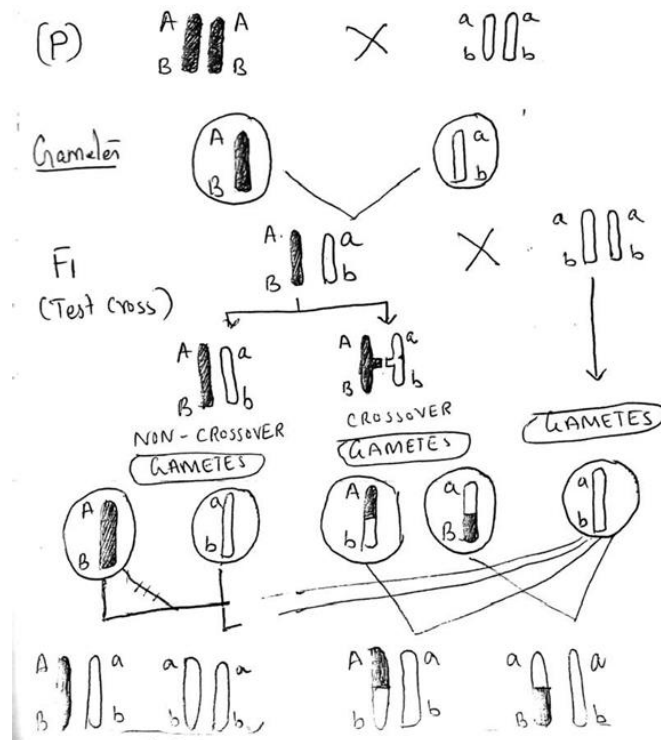
1. **COUPLING:** It is defined as tendency of allele either dominant or Recessive coming from same parent enter the same gametes in highest proportion and get inherited together.
2. **REPULSION:** It is defined as tendency of alleles either dominant or recessive coming from different parents to repel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

The concept of linkage, proposed by Morgan, 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 chromosome together constitute 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

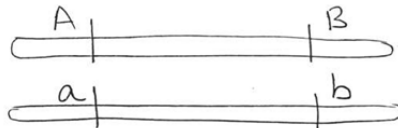




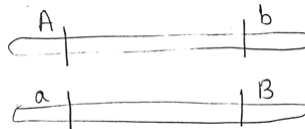
**Arrangement of Linked Genes:**

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

- a) **Cis - arrangement:** In this type, in the given pair of linked genes, two dominant alleles are located on one homologue of chromosomal pair and their recessive allele present on other homologous chromosome. Such heterozygotes are called Cis - 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 heterozygotes 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 characters 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 do not separate from each other to form new character complete linkage is very rare.

**Example:**

In wild drosophila, the gene 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.

<b>Parental generation</b>	<b>Grey vestigial</b>	<b>X</b>	<b>Black long (Ebony)</b>
<b>Genotype</b>	b <sup>+</sup> b <sup>+</sup> Vg Vg	X	bb Vg <sup>+</sup> Vg <sup>+</sup>
<b>Gametes</b>	(b <sup>+</sup> ) (Vg)		(b) (Vg <sup>+</sup> )
<b>F<sub>1</sub></b>	b <sup>+</sup> b Vg <sup>+</sup> Vg	→	Grey long

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

$$\begin{array}{l}
 \text{Test Cross} \quad b^1 b Vg^+ Vg \quad \times \quad bb Vg Vg \\
 \text{F}_1 \text{ Grey long} \quad \quad \quad \quad \quad \quad \quad \quad \text{Black vestigial} \\
 \\
 \text{Gametes} \quad (b^+ Vg) (b Vg^+) \quad (b Vg)
 \end{array}$$

**PUNNETT SQUARE**

	b Vg	Phenotype
b <sup>+</sup> Vg	b <sup>+</sup> b Vg Vg	Grey vestigial
b Vg <sup>+</sup>	bb Vg <sup>+</sup> Vg	black long

**Incomplete Linkage:**

In majority of organism, the linkage is incomplete. New characters appear in the off spring due to crossing over between homologous chromosomes. Hutchinson discovered incomplete linkage on endosperm characters in Maize.


**Example:**

For coupling Phase

- ✓ Colored aleurone (C) is dominant
- ✓ Colorless aleurone (c) is recessive
- ✓ Full endosperm (S) is dominant
- ✓ Shrunken endosperm (s) is recessive

Parental	Colored full		Colorless shrunken
Generation	C C S S	X	c c s s
Gametes	(C) (S)		(c) (s)
F <sub>1</sub> Generation	Cc Ss		coloured full
Test cross	Cc Ss	X	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) =  $\frac{180}{400} \times 100 = 90\%$

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

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 <sub>1</sub>	C c Ss		Coloured full
Test cross	Cc Sc	X	c c s s

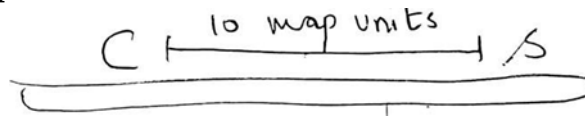
	cs	Phenotype	Expected Test cross ratio	Observed data out of 450
CS	Cc Ss	Colored full	100	20
Cs	Ccss	Colored Shrunken	100	180
cS	ccSs	Colorless full	100	180
cs	ccss	Colorless Shrunken	100	20
			1:1:1:1	400

Linkage & Recombination frequencies as for total population are

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

Non-parental charectors (cross over) =  $\frac{20+20}{400} \times 100 = 10\%$



**Chromosomal map:**

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

**CROSSING OVER**

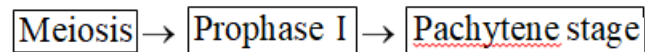
- ▲ This was first discovered by T.H. Morgan and castle in 1912. When linked genes tend to get separated and make new combinations. This type of mechanism are called CROKSSING, OVER
- ▲ The salient features of crossing over are
  - crossing over is an alternative to linkage
  - It separates linked genes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

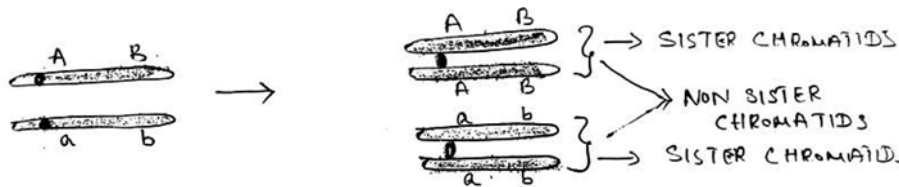


Crossing over occurs in the slage,

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.

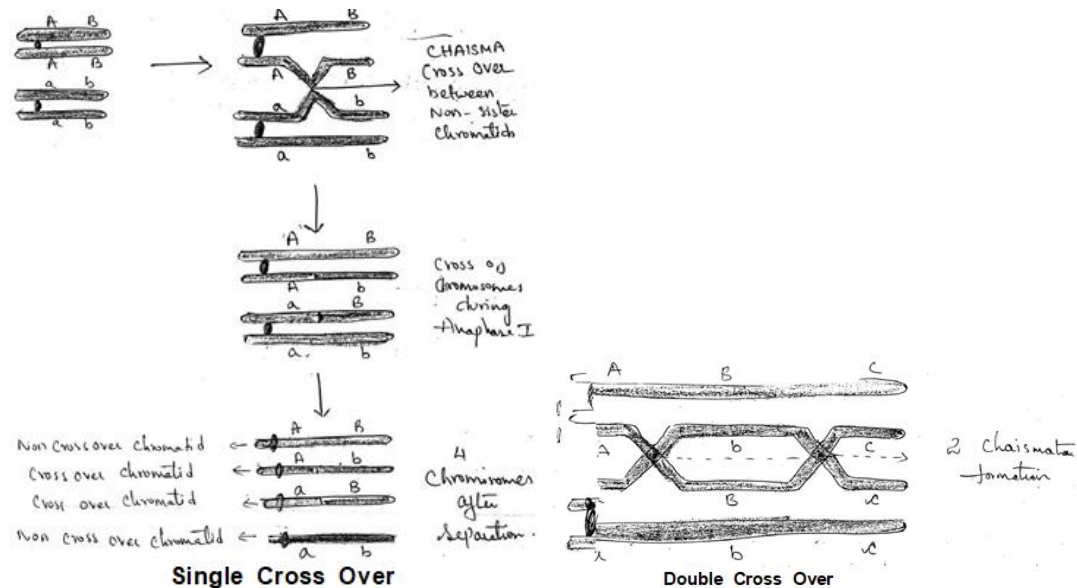
**Crossing Over:**

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



1. Crossing over 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. Number 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.

### Chiasma cross over



### CYTOLOGICAL EVIDENCES OF CROSSING OVER

Crossing over was proposed by T.H.Morgan in 1914, but it was clearly demonstrated by a cytological in 1931. Because of following reasons. Homologous chromosomes exchange reciprocal segments of chromatids.

1. Chromosomes after exchange don't show morphological 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 revered X and translocated X ch along.

**Types of X - chromosome**

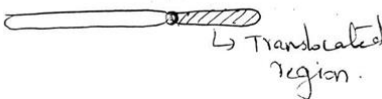
i) Normal X chromosome. (X)



ii) Short X'



i) Carrying Translocation. (X'')

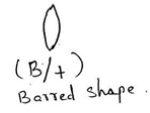


**Local Under Study in drosophila**

Eye shape

Normal wild

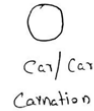
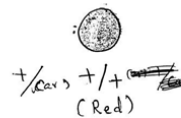
Mutant type



Eye Colour

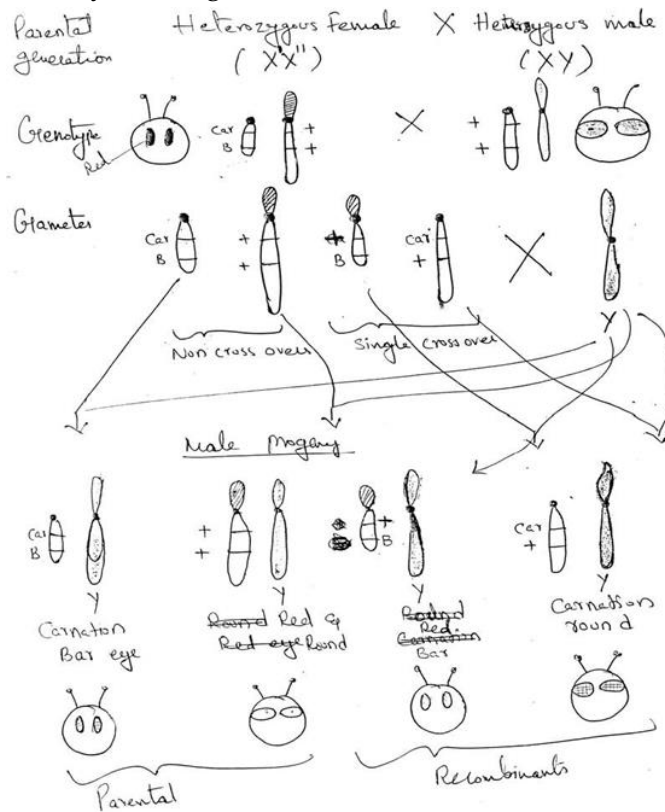
Normal wild

Mutant



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







P: Cc Bb X cc bb  
 Gamete (CB) (Cb) (c) (b)  
 (cB) (cb)

**Punnett square**

	Cb	Phenotype	Phenotype appearance
CB	CcBb	Red colour Bar eye	
Cb	Ccbb	Red colour Round eye	
cB	ccBb	Carnation color Bar eye	
cb	Ccbb	Carnation color Round eye	

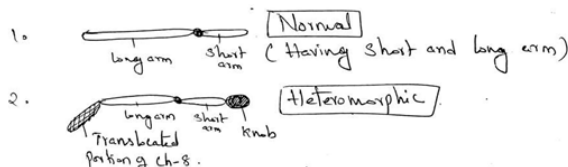
4 types of X - chromosomes are observed in all the four types of male flies.

- 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.**



Plants are crossed to check cytological proof.

P: Homozygous Heteromorphic X Homozygous Normal.  
 Genotype CCWW X ccww  
 Gametes (C) (W) (c) (w)  
 F<sub>1</sub> CcWw

**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
  - The value of RF (Z-value) is determined as 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 progeny &  $a_1$  and  $a_4$  are parental.  
g.: (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.

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.  
g.: (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.

If checking the Z value table, the percentage recombination for coupling 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 crossover % of recombinants.
- 3) Crossover % is converted into map distance which is expressed in map units or centimorgan units (cM), i.e.



1% of crossing over = 1 map units

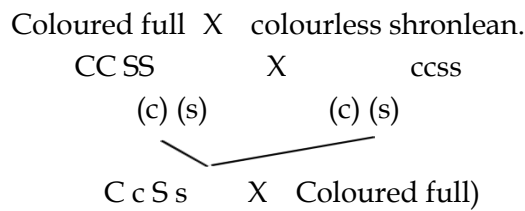
chromosomal map is also called as linkage map, genetic map.

These are two types of test crosses used to 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 aleurone and full endosperm (CCSS) crossed with excessive colourless aleurone and shrunken endosperm. (ccss)



**Test cross :** C c S s X cc ss

(CS) (Cs) (cs)

(cS) (cs)

Four types of offspring are produced in the following ratio.

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

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

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

$$= 96.4\%$$

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

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

$$= 3.6\%$$

∴ The chromosomal map is



**Three Test Cross**

colored aleuron full endosperm & Starchy endosperm		Colorless aleurone shrunken endosperm waxy endosperm
CCSSWW	X	ccssww
(C) (S) (W)	X	(c) (s) (w)
GEN Cs Sc Ww	=	(colored, full, starchy)
F1x F1 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	csW	csw

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
<b>Total</b>	<b>1000</b>	<b>100</b>	

i) Crossing over % b/w C-S

$$= \frac{\text{single crossover}(C -S) + \text{Double crossovers}}{\text{TotalNo.of offspring}} \times 100$$

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

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

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

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

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

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

iii) Cross over % b/w C-W

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

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

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

∴ 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 Sturtevant, 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 segregation of genes in F<sub>2</sub> generation such effect is called maternal effect.

- 1) The genes showing Non mendelian 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.

<b>Example -1:</b>	Female ( )	Male ( )
	Dextral	Sinistral
	Right coil	Left coil }Parent
	DD	dd

D-Gene produces protein & makes dextral

**Dd**  
(Dextral)

←Ddx Dd→ F<sub>1</sub>

<b>Dextral</b>	<b>Dextral</b>	
D	d	
D DD	Dd	→F <sub>2</sub>
Dextral	Dextral	
d Dd	dd	
Dextral	Dextral	

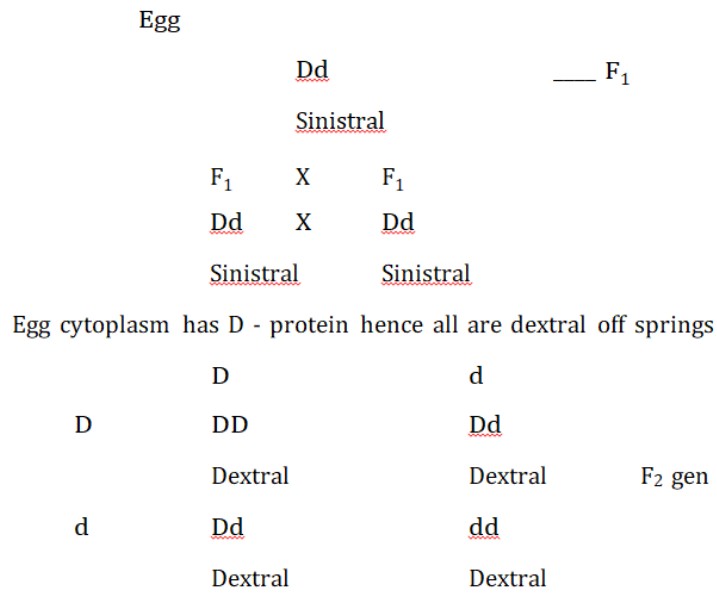
All off spring in F<sub>2</sub> generation shows dextral right coil because mothers cytoplasm provides D-gene with D-protein.

### Example - 2 :

<b>Female ( )</b>	<b>Male ( )</b>	
Sinistral	Dextral	Parental gen
Left Coil	Right coil	
dd	DD	
No Protein	(D) Gametes	

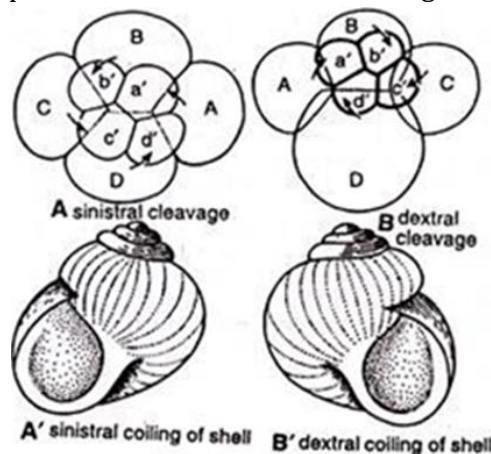


∴ sinistral

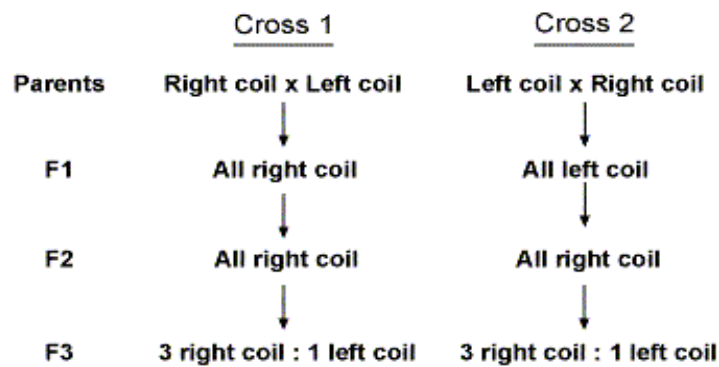


**Shell coiling in Snail and Maternal effects**

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.



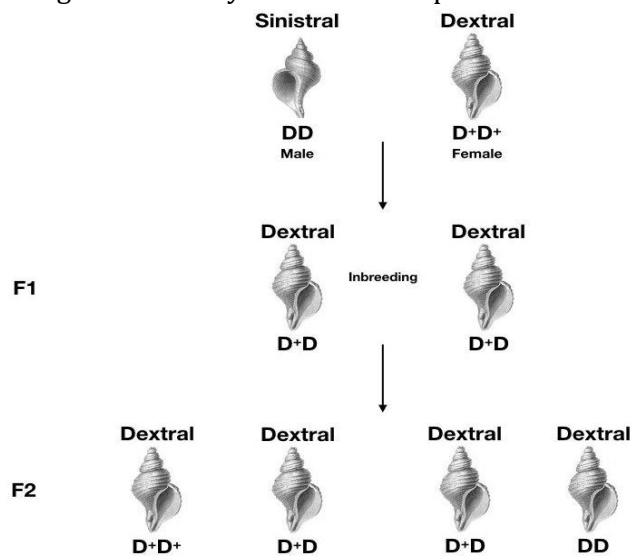
Coiling snail shells are a typical trait that shows maternal impact. The mother's genotype determines the coiling phenotype that the kids display. The outcomes of the following crosses between pure line snails were observed. It is customary to offer the female first.



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, pale & variegation plants would be obtained.

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



F<sub>1</sub> is green plants

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



F<sub>1</sub> is pale plants only

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



F<sub>1</sub> is green, pale & variegated plants.

**VARIATION IN LEAVES OF MIRABILIS 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.

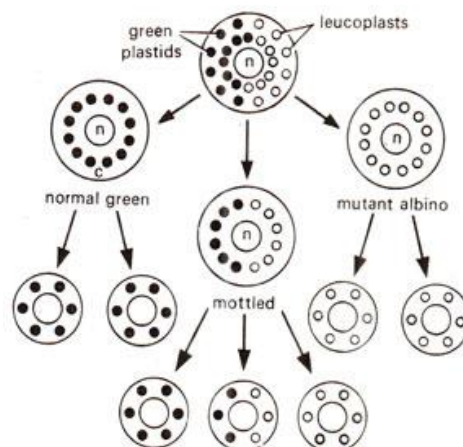
- 1. i) Cells with chloroplast Green (G)
- 2. ii) Cells with leucoplast Pala green (p)
- 3. iii) Cells with chloroplasts leucoplasts & Variegation (v)

**Example - 1:**

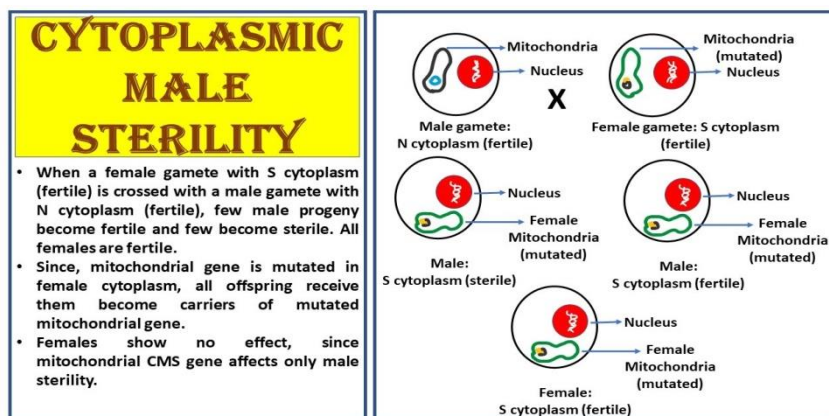
G - Green leaf

P - Pale Green

v - Variegation leaf



### 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 transitional 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 (carmin, lactophenol or iodine).

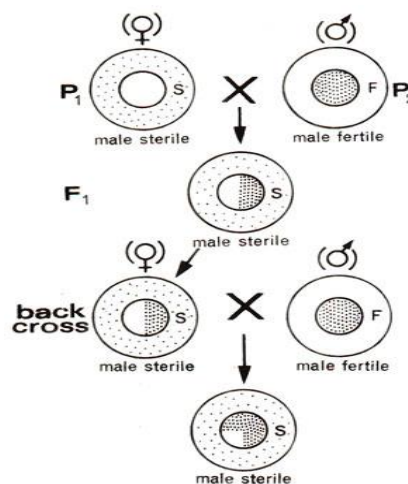
#### Genetic sterility



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.



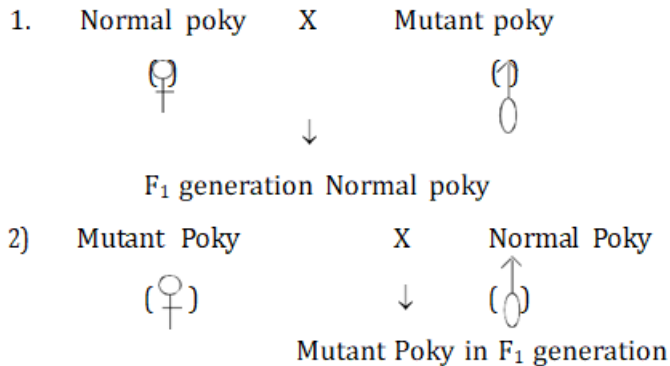
#### **4.4 The Poky Strain in Neurospora Crassa**

- ◆ 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 phosphorytation. i.e. ATP production.
- ◆ But in poky strains, these mitochondrial 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.



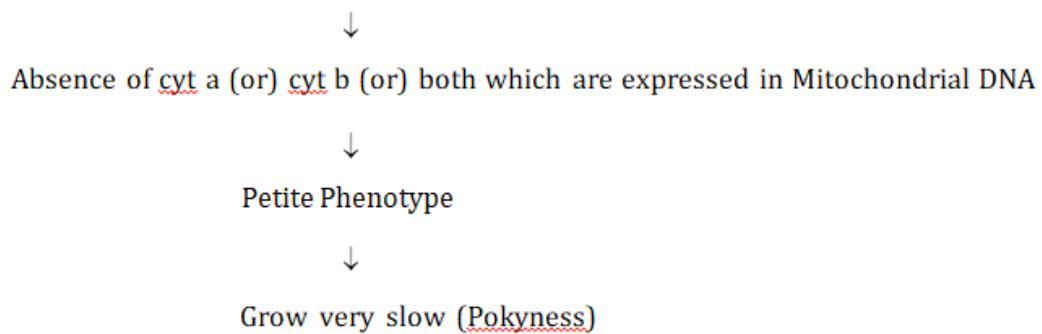
- ◆ i.e. one mutant poky strain is deficient in only cyt a while other is deficient in cyt b. These strains are called as mutant poky strain, they grow slow.

**Example for Non-Mendelian Inheritance:**



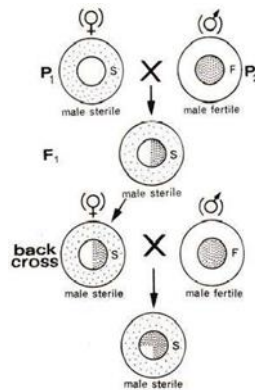
- ◆ Hence male gametes in Neurospora contributes negligible amount of cytoplasm just like higher animals & plants.
- ◆ Only female cytoplasm is inherited in progeny i.e.
- ◆ The factor for pokyness is resided in the female cytoplasm.
- ◆ This non-mendelian uniparental inheritance suggested that the cytoplasm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

Poky Mutant of Neurospora crassa



**LIFE CYCLE OF NEUROSPORA CRASSA :**

Cytoplasmic Male Sterility in Maize (Zea Mays) Introduction:

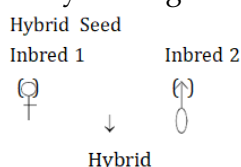




- ◆ Cytoplasmic male sterility (CMS) is the failure of plants to produce functional anthers, pollen or male gametes
- ◆ It shows Non - Mendelian inheritance, with male sterility inherited maternally. 2 types of cytoplasm.
  1. N (Normal )
  2. S (aberrant) is sterile
- ◆ CMS is important in hybrid maize 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 vigor.



### Different ways for Male sterility

1. Emasculation
2. CMS
3. Genetic male sterility (ms 1 to ms 52)
  - i) CMS - T → T - (Mutation 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 mitochondria and chloroplasts had their own genomes.

### Homoplasmy & Heteroplasmy

The number of copies of organelle genus (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 and said to exhibit homoplasmy.
- ◆ A cell or organism in which not all copies of an organelle gene are same is called Heteroplasmic & said to exhibit Heteroplasmy.

### Explain chloroplast inheritance of leaf - color phenotypes.

i) Egg cell ( ) X pollen ( )

If contains chloroplast / leukoplast or variegated, H is passed into the F1 progeny. This is because, Mother Cytoplasm is contributed to progeny not father's cytoplasm.

### A chloroplast Mutant in Chlamydomonas streptomycin Resistance

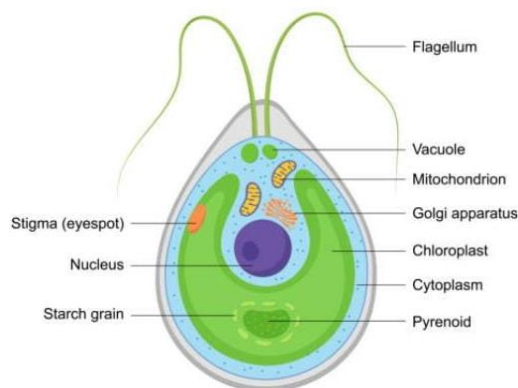
- ◆ The 1st Mutation in chlamydomonas was discovered by Ruth Sagar (1954) and confers streptomycin resistance (SmR) During mating of streptomycin resistant (SmR) female clamydomonas with streptomycin sensitive (Sms) allele, the F1 gen observed was stnR, this is because maternal inheritance observed in progeny.

### Diagram:

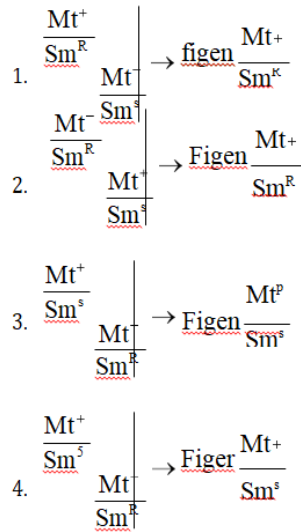
- ◆ Always Mt+ (mating type) cells one capable of mating while mt- are incapable.
- ◆ In this case, mother smR when crossed with Sm3 progeny observed were Sm<sup>R</sup>
- ◆ In this example, Mother is mt<sup>+</sup> & sm<sup>5</sup> when crossed with father mt<sup>-</sup> & sm<sup>R</sup> the F<sub>1</sub> gen observed where mt<sup>+</sup> sm<sup>5</sup>.
- ◆ Cytoplasmic inheritance is inherited from mother.

### STRUCTURE OF CHLAMYDOMONAS REINHARDTII

#### CHLAMYDOMONAS



**Mutant Chlamydomonas**



**4.6 HARDY - WEINBERG EQUILIBRIUM (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 disruptive 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:** Disrupts allele frequencies.

**Natural Selection:** Disrupts gene frequencies.

**Non-Random mating:** Also disrupts gene frequencies.

**Genetic drift:** Disrupts allele frequencies.

**Gene Flow:** Disrupts allele frequencies.

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

**Hardy - Weinberg Principle**

Parental generation

**Phenotype**                      YY                      Yg                      yy

<b>Genotypic frequency</b>	0.49	0.42	0.09
<b>No. of Individuals(500) }</b>	245	210	45
No of alleles Y :	245 + 245		y: 45 + 45
(Total : 1000) =	490 + 210		: 90 + 210
	= 700		= 300
<b>Allelic frequency }</b>	$\frac{700}{700+300}$	$\frac{300}{700+300}$	
	= 0.7	= 0.3	
	= p	= q	

Hardy weinberg Analysis for genotype frequency.

	p(0.7)	q (0.3)
	Y	y
0.7	YY	<u>Yy</u>
P Y	P <sup>2</sup> = 0.49	<u>pq</u> = 0.21
0.3q y	<u>Yy</u> pq=0.21	<u>yy</u> q <sup>2</sup> = 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 recessive genotype (aa) is 36%, using this percentage calculate the following.

- The genotypic frequency of "aa"?  
Ans: 36%
- The allelic frequency of "a"?  
Ans: The frequency of aa = 36%  
i.e.,  $q^2 = 0.36$   
then  $q = 0.6$   
allelic frequency of a = 60%
- The allelic frequency of "A"?  $p + q = 1$ ,  $q = 0.6$   
 $p = 1 - 0.6$   
 $p = 0.4$   
frequency of A is 40%
- What is the frequency of AA and Aa?  
 $AA = p^2$  ( $p = 0.4$ )  
 $= 0.4 \times 0.4$



$$= 0.16$$

frequency of AA is 16%  $Aa = 2pq$

$$= 2 \cdot 0.4 \cdot 0.6$$

$$= 0.48$$

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

AAAa

$$\text{Frequency} = 16\% + 48\%$$

**Problem -2:**

A very large population of randomly mating laboratory mice contains 35% white mice. White color is caused by homozygous recessive genotype (aa). Calculate allelic & genotypic frequencies?

**Sol:**

Homozygous recessive genotype = aa

According to allelic frequency formula

$$p + q = 1, \text{ where } q = a$$

$$\therefore q^2 = aa$$

$$q^2 = 0.35 \quad (35 \times 100)$$

$$\text{Hence } q = \sqrt{0.35}$$

$$q = 0.59$$

$$p + q = 1$$

$$p = 1 - q$$

$$p = 1 - 0.59$$

$$= 0.41$$



According to genotypic frequency

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

Where (i)  $p = 0.41$

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

$$\text{ii) } 2pq = 2 \times 0.41 \times 0.59$$

$$= 0.48 \rightarrow (2)$$

$$\text{iii) } q^2 = 0.59 \times 0.59$$

$$= 0.35 \rightarrow (3)$$

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

$$0.17 + 0.48 + 0.35 = 1$$

Hence the given observation satisfies Hardy weinberg equilibrium.

### Hardy-Weinberg Equations

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

$p$  = frequency of the dominant allele  
in a population

$$p + q = 1$$

$q$  = frequency of the recessive allele  
in a population

