



PITHAPUR RAJAH'S GOVERNMENT COLLEGE

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cell biology and genetics

Cell and Cell Organelles:

Introduction- Higher eukaryotes have multiple organs to perform specific functions such as liver, kidney and heart. Each Organ has specific tissue and each tissue is composed of cells. **“Cell is the structural and functional unit of life”** and it contains all necessary infrastructure to perform all functions. Based on cellular structure, cells are classified as prokaryotic and eukaryotic cells. In most of the cases, prokaryotes are single cells where as eukaryotes are either single cells or part of multicellular tissues system. Besides this, both types of cells have several structural and metabolic differences as given in Table 3.1 and are discussed later in the lecture.

Feature	Prokaryote	Eukaryote
Size	Small, in μm range	Variable size, upto $40\mu\text{m}$ in diameter.
Genetic material	Circular DNA present in cytosol as free material	DNA in the form of linear chromosome present in well defined double membrane nucleus, no direct connection with cytosol
Replication	Single origin of replication	Multiple origin of replication.
Genes	No Intron	Presence of Intron
Organelles	No membrane bound organelles	Membrane bound organelles with well defined function.
Cell walls	Very complex cell wall	Except Fungi and plant, eukaryotic cells are devoid of a thick cell wall.
Ribosome	70S	80S
Transcription and translation	Occurs together	Transcription in nucleus and translation in cytosol

Prokaryotic and Eukaryotic cells

The cell was first seen by Robert Hooke in 1665 using a primitive, compound microscope. He observed very thin slices of cork and saw a multitude of tiny structures that he resembled to walled compartments of a monk. Hence, named them cells. Hooke's description of these cells was published in Micrographia. The cell is smallest unit of a living system and fall in the microscopic range of 1 to $100\mu\text{m}$. They attain various shapes and sizes to attain variety of functions. The understanding of cell is necessary to understand the structure and function of a living organism. One of most important

characteristics of cell is ability to divide. The existence of a cell indicates that it has evolved from an already existing cell and further it can give rise to a new cell. This was first stated by Theodor Schwann. Pioneering work by Theodor Schwann, Matthias Jakob Schleiden on cells, gave birth to the cell theory. Their theory states:

1. All living things are made of cells.
2. Cells are the basic building units of life.
3. New cells are created by old cells dividing into two.

In 1855, Rudolf Virchow added another point to the theory and concluded that all cells come from pre-existing cells, thus completing the classical cell theory. The cell theory holds true for all living things, no matter how big or small, or how simple or complex. Viruses are exception to the cell theory. Cells are common to all living beings, and provide information about all forms of life. Because all cells come from existing cells, scientists can study cells to learn about growth, reproduction, and all other functions that living things perform. By learning about cells and how they function, we can learn about all types of living things.

Classification of cells:

Classification of cells:

Any living organism may contain only one type of cell either **A. Prokaryotic cells**; **B. Eukaryotic cells**. The terms prokaryotic and eukaryotic were suggested by Hans Ris in the 1960's. This classification is based on their complexity. Further based on the kingdom into which they may fall i.e the plant or the animal kingdom, plant and animal cells bear many differences. These will be studied in detail in the upcoming sections.

Prokaryotic cells

Prokaryote means before nucleus in Greek. They include all cells which lack nucleus and other membrane bound organelles. Mycoplasma, virus, bacteria and cyanobacteria or blue-green algae are prokaryotes.

Most prokaryotes range between 1 μm to 10 μm , but they can vary in size from 0.2 μm to 750 μm (*Thiomargarita namibiensis*). They belong to two taxonomic domains which are the bacteria and the archaea. Most prokaryotes are unicellular, exceptions being myxobacteria which have multicellular stages in their life cycles. They are membrane bound mostly unicellular organisms lacking any

internal membrane bound organelles. A typical prokaryotic cell is schematically illustrated in Figure 1. Though prokaryotes lack cell organelles they harbor few internal structures, such as the cytoskeletons, ribosomes, which translate mRNA to proteins. Membranous organelles are known in some groups of prokaryotes, such as vacuoles or membrane systems devoted to special metabolic properties, e.g., photosynthesis or chemolithotrophy. In addition, some species also contain protein-enclosed microcompartments, which have distinct physiological roles (carboxysomes or gas vacuoles).

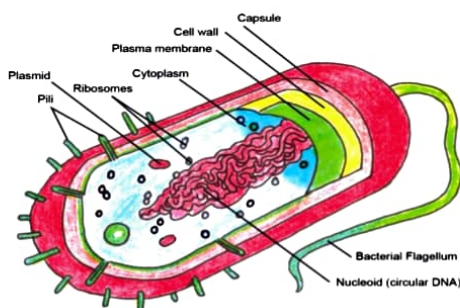


Figure 1: Schematic diagram of a prokaryotic cell

The individual structures depicted in Figure 1 are as follows and details will be discussed in forthcoming chapters:

Flagella: It is a long, whip-like protrusion found in most prokaryotes that aids in cellular locomotion. Besides its main function of locomotion it also often functions as a sensory organelle, being sensitive to chemicals and temperatures outside the cell.

Capsule: The capsule is found in some bacterial cells, this additional outer covering protects the cell when it is engulfed by phagocytes and by viruses, assists in retaining moisture, and helps the cell adhere to surfaces and nutrients. The capsule is found most commonly among Gram-negative bacteria. *Escherichia coli*, *Klebsiella pneumoniae*, *Haemophilus influenzae*, *Pseudomonas aeruginosa* and *Salmonella* are some examples Gram-negative bacteria possessing capsules. Whereas examples

Cell membrane: Cell membrane surrounds the cell's cytoplasm and regulates the flow of substances in and out of the cell. It will be discussed in detail in one of the coming chapters.

Cytoplasm: The cytoplasm of a cell is a fluid in nature that fills the cell and is composed mainly of 80% water that also contains enzymes, salts, cell organelles, and various organic molecules. The details will be discussed in forthcoming chapter.

Ribosomes: Ribosomes are the organelles of the cell responsible for protein synthesis. Details of ribosomes will be explained in coming chapter.

Nucleoid Region: The nucleoid region is possessed by a prokaryotic bacterial cell. It is the area of the cytoplasm that contains the bacterial DNA molecule.

Plasmids: The term plasmid was first introduced by the American molecular biologist Joshua Lederberg in 1952. A plasmid is a DNA molecule (mostly in bacteria) that is separate from, and can replicate independently of, the chromosomal DNA. They are double-stranded and circular. Plasmids usually occur naturally in bacteria, but are sometimes found in eukaryotic organisms. Their sizes vary from 1 to over 1,000 kbp. The number of identical plasmids in a single cell can range anywhere from one to thousands under some circumstances and it is represented by the copy number. Plasmids can be considered mobile because they are often associated with conjugation, a mechanism of horizontal gene transfer. Plasmids that can coexist within a bacterium are said to be compatible. Plasmids which cannot coexist are said to be incompatible and after a few generations are lost from the cell. Plasmids that encode their own transfer between bacteria are termed conjugative. Non-conjugative plasmids do not have these transfer genes but can be carried along by conjugative plasmids via a mobilisation site. Functionally they carry genes that code for a wide range of metabolic activities, enabling their host bacteria to degrade pollutant compounds, and produce antibacterial proteins. They can also harbour genes for virulence that help to increase pathogenicity of bacteria causing diseases such as plague, dysentery, anthrax and tetanus. They are also responsible for the spread of antibiotic resistance genes that ultimately have an impact on the treatment of diseases. Plasmids are classified into the following types.

Pili: Pili are hair-like structures on the surface of the cell that help attach to other bacterial cells. Shorter pili called fimbriae help bacteria attach to various surfaces. A pilus is typically 6 to 7 nm in diameter. The types of pili are Conjugative pili and Type IV pili. Conjugative pili allow the transfer of DNA between bacteria, in the process of bacterial conjugation. Some pili, called type IV pili, generate motile forces.

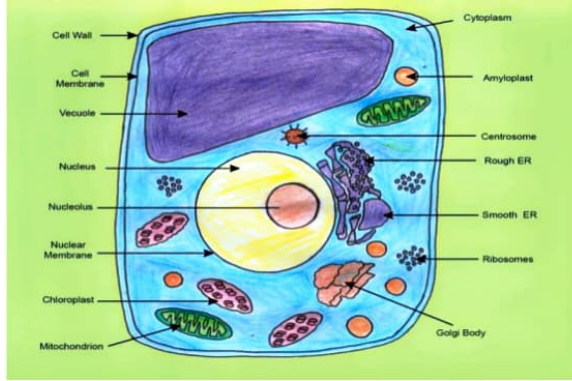
Morphology of prokaryotic cells

Prokaryotic cells have various shapes; the four basic shapes are (Figure 3):

- Cocci - spherical
- Bacilli - rod-shaped
- Spirochaete - spiral-shaped
- Vibrio - comma-shaped

Reproduction

Bacteria and archaea reproduce through asexual reproduction known as binary fission. Binary fission is an asexual mode of reproduction. During binary fission, the genomic DNA undergoes replication and the original cell is divided into two identical cells. Due to binary fission, all organisms in a colony are genetically equivalent (Figure 4). The process begins with DNA replication followed by DNA segregation, division site selection, invagination of the cell envelope and synthesis of new cell

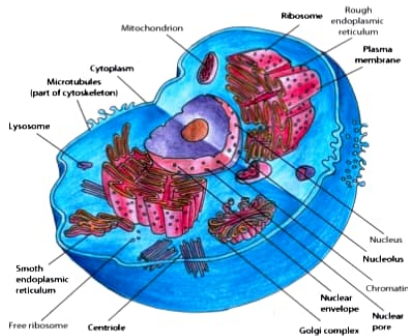


Schematic representation of a plant cell.

Animal cells:

An animal cell is a form of eukaryotic cell that makes up many tissues in animals. Figure 7 depicts a typical animal cell. The animal cell is distinct from other eukaryotes, most notably plant cells, as they

lack cell walls and chloroplasts, and they have smaller vacuoles. Due to the lack of a rigid cell wall, animal cells can adopt a variety of shapes, and a phagocytic cell can even engulf other structures. There are many different cell types. For instance, there are approximately 210 distinct cell types in the adult human body.



Schematic representation of a typical animal cell.

Cell organelles in animal cell:

Cell membrane: Plasma membrane is the thin layer of protein and fat that surrounds the cell, but is inside the cell wall. The cell membrane is semipermeable, allowing selective substances to pass into the cell and blocking others.

Nucleus: They are spherical body containing many organelles, including the nucleolus. The nucleus controls many of the functions of the cell (by controlling protein synthesis) and contains DNA (in chromosomes). The nucleus is surrounded by the nuclear membrane and possesses the nucleolus which is an organelle within the nucleus - it is where ribosomal RNA is produced.

Golgi apparatus: It is a flattened, layered, sac-like organelle involved in packaging proteins and carbohydrates into membrane-bound vesicles for export from the cell.

Ribosome and Endoplasmic reticulum: Ribosomes are small organelles composed of RNA-rich cytoplasmic granules that are sites of protein synthesis and Endoplasmic reticulum are the sites of

a. Rough endoplasmic reticulum: These are a vast system of interconnected, membranous, infolded and convoluted sacks that are located in the cell's cytoplasm (the ER is continuous with the outer nuclear membrane). Rough ER is covered with ribosomes that give it a rough appearance. Rough ER transport materials through the cell and produces proteins in sacks called cisternae (which are sent to the Golgi body, or inserted into the cell membrane).

b. Smooth endoplasmic reticulum: These are a vast system of interconnected, membranous, infolded and convoluted tubes that are located in the cell's cytoplasm (the ER is continuous with the outer nuclear membrane). The space within the ER is called the ER lumen. Smooth ER transport materials through the cell. It contains enzymes and produces and digests lipids (fats) and membrane proteins; smooth ER buds off from rough ER, moving the newly-made proteins and lipids to the Golgi body and membranes.

Mitochondria: These are spherical to rod-shaped organelles with a double membrane. The inner membrane is infolded many times, forming a series of projections (called cristae). The mitochondrion converts the energy stored in glucose into ATP (adenosine triphosphate) for the cell.

Lysosome: Lysosomes are cellular organelles that contain the hydrolase enzymes which breaks down waste materials and cellular debris. They can be described as the stomach of the cell. They are found in animal cells, while in yeast and plants the same roles are performed by lytic vacuoles. Lysosomes digest excess or worn-out organelles, food particles, and engulf viruses or bacteria. The membrane around a lysosome allows the digestive enzymes to work at the 4.5 pH they require. Lysosomes fuse with vacuoles and dispense their enzymes into the vacuoles, digesting their contents. They are created by the addition of hydrolytic enzymes to early endosomes from the Golgi apparatus.

Centrosome: They are small body located near the nucleus and has a dense center and radiating tubules. The centrosomes are the destination where microtubules are made. During mitosis, the centrosome divides and the two parts move to opposite sides of the dividing cell. Unlike the centrosomes in animal cells, plant cell centrosomes do not have centrioles.

Peroxisome

Peroxisomes are organelles that contain oxidative enzymes, such as D-amino acid oxidase, ureate oxidase, and catalase. They may resemble a lysosome, however, they are not formed in the Golgi complex. Peroxisomes are distinguished by a crystalline structure inside a sac which also contains amorphous gray material. They are self replicating, like the mitochondria. Components accumulate at a given site and they can be assembled into a peroxisome. Peroxisomes function to rid the body of toxic substances like hydrogen peroxide, or other metabolites. They are a major site of oxygen utilization and are numerous in the liver where toxic byproducts accumulate.

Vacuoles and vesicles

Vacuoles are single-membrane organelles that are essentially part of the outside that is located within the cell. The single membrane is known in plant cells as a tonoplast. Many organisms will use vacuoles as storage areas. Vesicles are much smaller than vacuoles and function in transporting materials both within and to the outside of the cell.

10.1.1 Phases of Cell Cycle

A typical eukaryotic cell cycle is illustrated by human cells in culture. These cells divide once in approximately every 24 hours (Figure 10.1). However, this duration of cell cycle can vary from organism to organism and also from cell type to cell type. Yeast for example, can progress through the cell cycle in only about 90 minutes.

The cell cycle is divided into two basic phases:

- Interphase
- M Phase (Mitosis phase)

The M Phase represents the phase when the actual cell division or mitosis occurs and the interphase represents the phase between two successive M phases. It is significant to note that in the 24 hour average duration of cell cycle of a human cell, cell division proper lasts for only about an hour. The interphase lasts more than 95% of the duration of cell cycle.

The M Phase starts with the nuclear division, corresponding to the separation of daughter chromosomes (karyokinesis) and usually ends with division of cytoplasm (cytokinesis). The interphase, though called the resting phase, is the time during which the cell is preparing for division by undergoing both cell growth and DNA replication in an orderly manner. The interphase is divided into three further phases:

- G₁ phase (Gap 1)
- S phase (Synthesis)
- G₂ phase (Gap 2)

G₁ phase corresponds to the interval between mitosis and initiation of DNA replication. During G₁ phase, the cell is metabolically active and continuously grows but does not replicate its DNA. S or **synthesis** phase marks the period during which DNA synthesis or replication takes place. During this time the amount of DNA per cell doubles. If the initial amount of DNA is denoted as 2C then it increases to 4C. However, there is no increase in the chromosome number; if the cell had diploid or 2n number of chromosomes at G₁, even after S phase the number of chromosomes remains the same, i.e., 2n.

In animal cells, during the S phase, DNA replication begins in the nucleus, and the centriole duplicates in the cytoplasm. During the G₂ phase, proteins are synthesised in preparation for mitosis while cell growth continues.

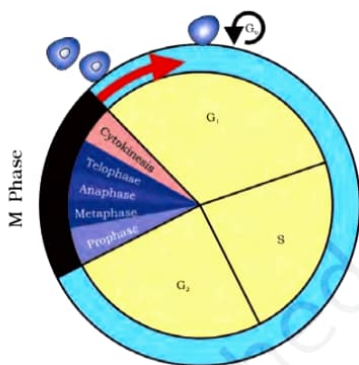


Figure 10.1 A diagrammatic view of cell cycle indicating formation of two cells from one cell

How do plants and animals continue to grow all their lives? Do all cells in a plant divide all the time? Do you think all cells continue to divide in all plants and animals? Can you tell the name and the location of tissues having cells that divide all their life in higher plants? Do animals have similar meristematic tissues?

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You have studied mitosis in onion root tip cells. It has 16 chromosomes in each cell. Can you tell how many chromosomes will the cell have at G₁ phase, after S phase, and after M phase? Also, what will be the DNA content of the cells at G₁, after S and at G₂, if the content after M phase is 2C?

Some cells in the adult animals do not appear to exhibit division (e.g., heart cells) and many other cells divide only occasionally, as needed to replace cells that have been lost because of injury or cell death. These cells that do not divide further exit G₁ phase to enter an inactive stage called **quiescent stage (G₀)** of the cell cycle. Cells in this stage remain metabolically active but no longer proliferate unless called on to do so depending on the requirement of the organism.

In animals, mitotic cell division is only seen in the diploid somatic cells. However, there are few exceptions to this where haploid cells divide by mitosis, for example, male honey bees. Against this, the plants can show mitotic divisions in both haploid and diploid cells. From your recollection of examples of alternation of generations in plants (Chapter 3) identify plant species and stages at which mitosis is seen in haploid cells.

10.2 M PHASE

This is the most dramatic period of the cell cycle, involving a major reorganisation of virtually all components of the cell. Since the number of chromosomes in the parent and progeny cells is the same, it is also called as **equational division**. Though for convenience mitosis has been divided into four stages of nuclear division (karyokinesis), it is very essential to

Also, what will be the DNA content of the cells at G_1 , after S and at G_2 , if the content after M phase is $2C$?

show mitotic divisions in both haploid and diploid cells. From your recollection of examples of alternation of generations in plants (Chapter 3) identify plant species and stages at which mitosis is seen in haploid cells.

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- **Prophase**
- **Metaphase**
- **Anaphase**
- **Telophase**

10.2.1 Prophase

Prophase which is the first stage of karyokinesis of mitosis follows the S and G_2 phases of interphase. In the S and G_2 phases, the new DNA molecules formed are not distinct but intertwined. Prophase is marked by the initiation of condensation of chromosomal material. The chromosomal material becomes untangled during the process of chromatin condensation (Figure 10.2 a). The centrosome, which had undergone duplication during S phase of interphase, now begins to move towards opposite poles of the cell. The completion of prophase can thus be marked by the following characteristic events:

- Chromosomal material condenses to form compact mitotic chromosomes. Chromosomes are seen to be composed of two chromatids attached together at the centromere.
- Centrosome which had undergone duplication during interphase, begins to move towards opposite poles of the cell. Each centrosome radiates out microtubules called asters. The two asters together with spindle fibres forms mitotic apparatus.

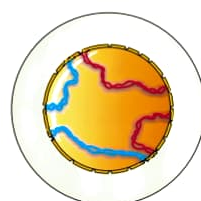
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Cells at the end of prophase, when viewed under the microscope, do not show golgi complexes, endoplasmic reticulum, nucleolus and the nuclear envelope.

10.2.2 Metaphase

The complete disintegration of the nuclear envelope marks the start of the second phase of mitosis, hence the chromosomes are spread through the cytoplasm of the cell. By this stage, condensation of chromosomes is completed and they can be observed clearly under the microscope. This then, is the stage at which morphology of chromosomes is most easily studied. At this stage, metaphase chromosome is made up of two sister chromatids, which are held together by the centromere (Figure 10.2 b). Small disc-shaped structures at the surface of the centromeres are called kinetochores. These structures serve as the sites of attachment of spindle fibres to the chromosomes that are moved into position at the centre of the cell. Hence, the metaphase is characterised by all the chromosomes coming to lie at the equator with one chromatid of each chromosome connected by its kinetochore to spindle fibres from one pole and its sister chromatid connected by its kinetochore to spindle fibres from the opposite pole (Figure 10.2 b). The plane of alignment of the chromosomes at metaphase is referred to as the **metaphase plate**. The key features of metaphase are:

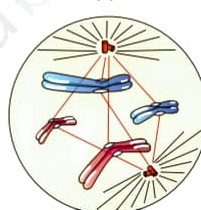
- Spindle fibres attach to kinetochores of chromosomes.
- Chromosomes are moved to spindle equator and get



Early Prophase



Late Prophase
(a)



Transition to
Metaphase

10.2.3 Anaphase

At the onset of anaphase, each chromosome arranged at the metaphase plate is split simultaneously and the two daughter chromatids, now referred to as daughter chromosomes of the future daughter nuclei, begin their migration towards the two opposite poles. As each chromosome moves away from the equatorial plate, the centromere of each chromosome remains directed towards the pole and hence at the leading edge, with the arms of the chromosome trailing behind (Figure 10.2 c). Thus, anaphase stage is characterised by

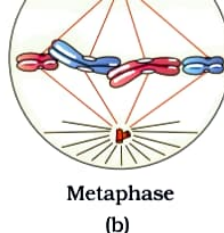
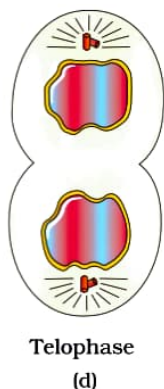
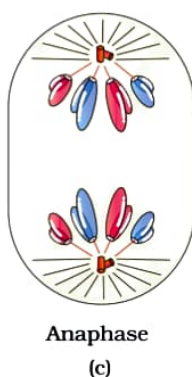


Figure 10.2 a and b : A diagrammatic view of stages in mitosis

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the following key events:

- Centromeres split and chromatids separate.
- Chromatids move to opposite poles.

10.2.4 Telophase

At the beginning of the final stage of karyokinesis, i.e., telophase, the chromosomes that have reached their respective poles decondense and lose their individuality. The individual chromosomes can no longer be seen and each set of chromatin material tends to collect at each of the two poles (Figure 10.2 d). This is the stage which shows the following key events:

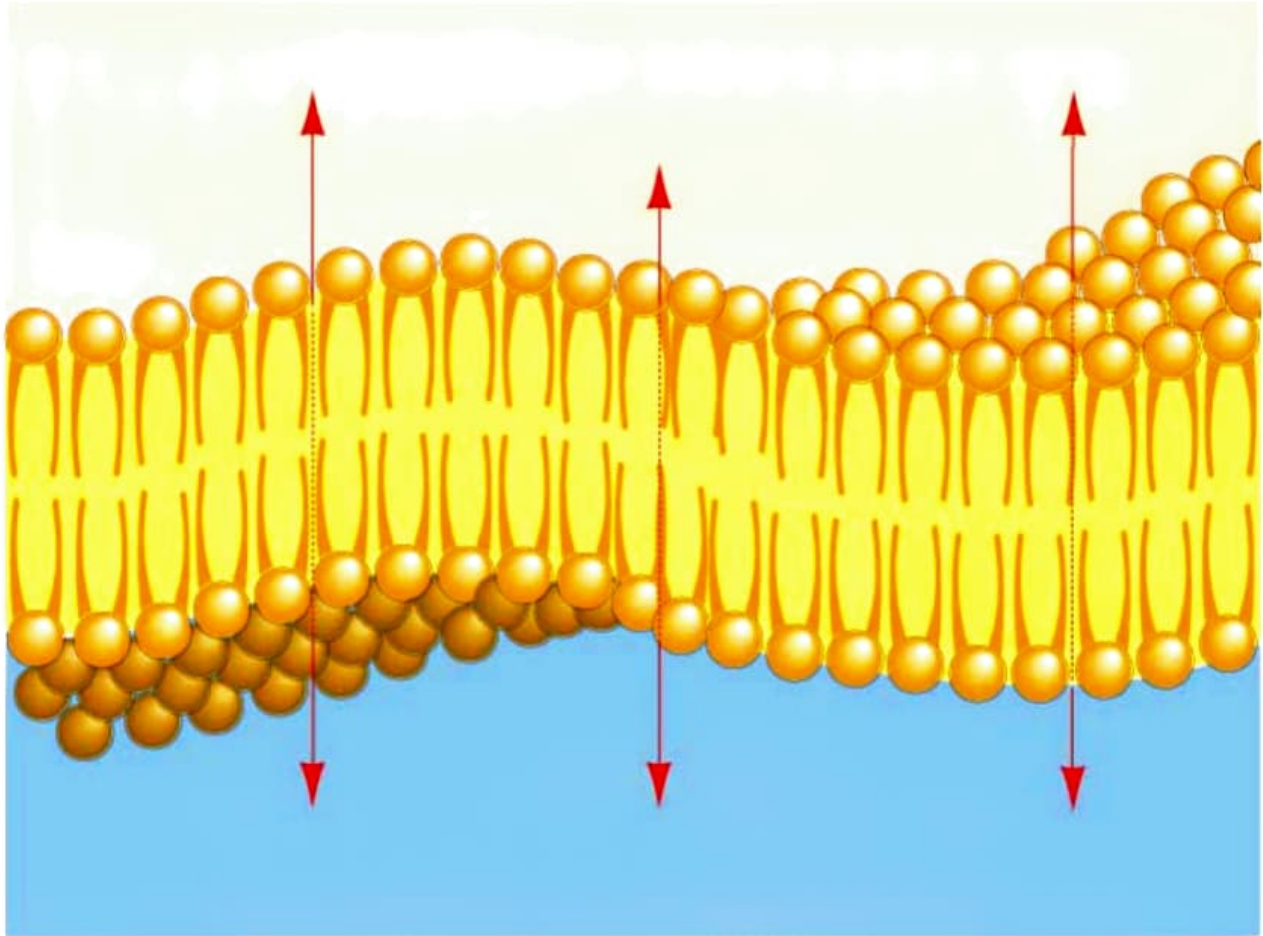
- Chromosomes cluster at opposite spindle poles and their identity is lost as discrete elements.
- Nuclear envelope develops around the chromosome clusters at each pole forming two daughter nuclei.
- Nucleolus, golgi complex and ER reform.

10.2.5 Cytokinesis

Mitosis accomplishes not only the segregation of duplicated chromosomes into daughter nuclei (karyokinesis), but the cell itself is divided into two daughter cells by the separation of cytoplasm called cytokinesis at the end of which cell division gets completed (Figure 10.2 e). In an animal cell, this is achieved by the appearance of a furrow in the plasma membrane. The furrow gradually deepens and ultimately joins in the centre dividing the cell cytoplasm into two. Plant cells however, are enclosed by a relatively inextensible cell wall, therefore they undergo cytokinesis by a different mechanism. In plant cells, wall formation starts in the centre of the cell and grows outward to meet the existing lateral walls. The formation of the new cell wall begins with the formation of a simple precursor, called the **cell-plate** that represents the middle lamella between the walls of two adjacent cells. At the time of cytoplasmic division, organelles like mitochondria and plastids get distributed between the two daughter cells. In some organisms karyokinesis is not followed by cytokinesis as a result of which multinucleate condition arises leading to the formation of syncytium (e.g., liquid endosperm in coconut).

Figure 10.2 c to e : A diagrammatic view of stages in Mitosis

Cell Membrane Structure and Function

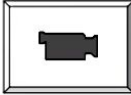


Chapter 4: Membrane Structure and Function

Plasma Membrane: Thin barrier separating inside of cell (cytoplasm) from outside environment

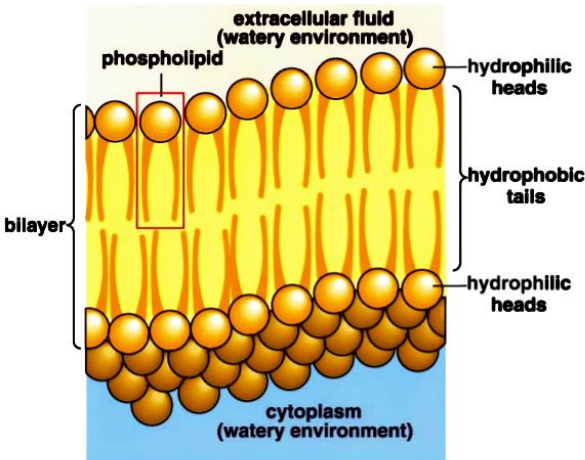
Function:

- 1) Isolate cell's contents from outside environment
- 2) Regulate exchange of substances between inside and outside of cell
- 3) Communicate with other cells

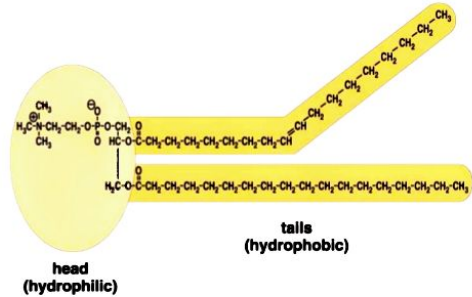


Phospholipid Bilayer: Double layer of phospholipids

- Hydrophilic ends form outer border
- Hydrophobic tails form inner layer



- Lipid tails of phospholipids are unsaturated ($C = C$)



Chapter 4: Membrane Structure and Function

Cell Membrane Proteins:

1) Transport Proteins:

- Regulate movement of hydrophilic molecules through membrane
 - A) Channel Proteins (e.g. Na^+ channels)
 - B) Carrier Proteins (e.g. glucose transporter)

2) Receptor Proteins:

- Trigger cell activity when molecule from outside environment binds to protein

3) Recognition Proteins:

- Allow cells to recognize / attach to one another
- Glycoproteins: Proteins with attached carbohydrate groups

Chapter 4: Membrane Structure and Function

How are Substances Transported Across Membranes?

Answer: Concentration Gradients

Concentration = Number of molecules in a given unit of volume (e.g. grams / liter; moles / liter)

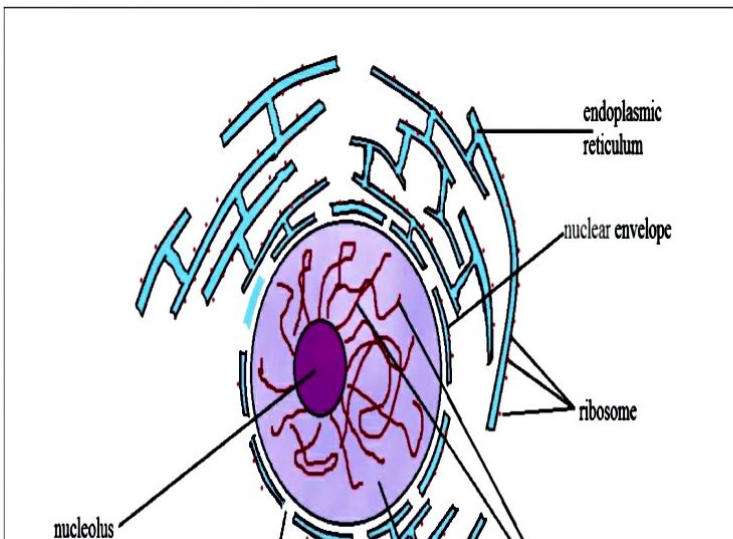
Nucleus:

- The cell nucleus is a membrane-bound structure that contains the cell's hereditary information and controls the cell's growth and reproduction.
- It is the command center of a eukaryotic cell and is commonly the most prominent organelle in a cell accounting for about 10 percent of the cell's volume.
- In general, a eukaryotic cell has only one nucleus. However, some eukaryotic cells are enucleated cells (without a nucleus), for example, red blood cells (RBCs); whereas, some are multinucleate (consists of two or more nuclei), for example, slime **molds**.
- The nuclear envelope has a complex structure consisting of:
 - a) Two nuclear membranes separated by a perinuclear space measuring about 20–40 nm.
 - b) Underlying nuclear lamina

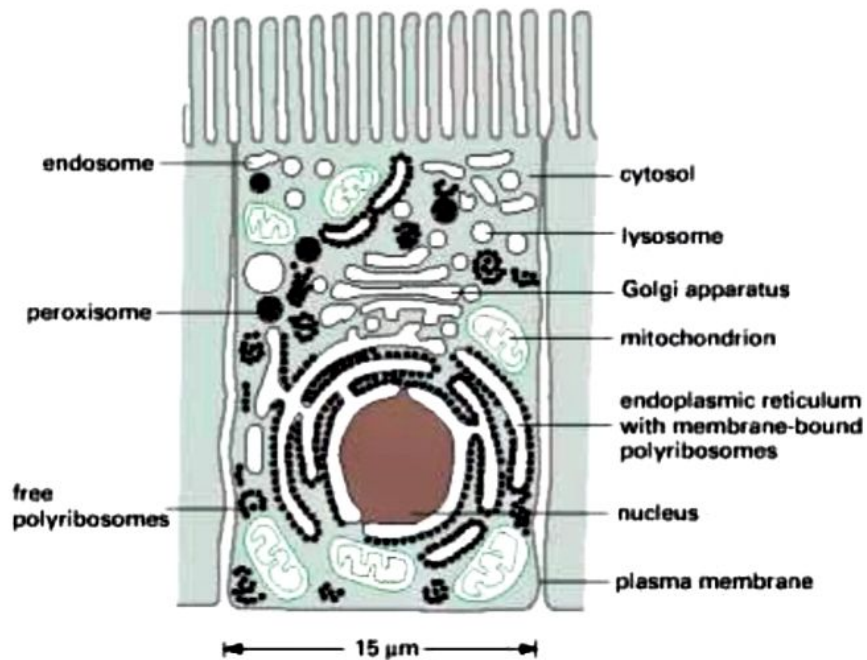
Nuclear membrane:

- The nucleus is surrounded by a system of two concentric membranes, called the inner and outer nuclear membranes
- The inner and outer nuclear membranes are joined at nuclear pore complex.
- The outer nuclear membrane is continuous with the endoplasmic reticulum, so the space between the inner and outer nuclear membranes is directly connected with the lumen of the ER. It is functionally similar to the membranes of the ER and has ribosomes bound to its cytoplasmic surface but protein composition differs slightly as they are enriched in proteins which binds to cytoskeleton
- The inner nuclear membrane carries proteins that are specific to the nucleus, such as those that bind the lamina

- The critical function of the nuclear membranes is to act as a barrier that separates the contents of the nucleus from the cytoplasm.



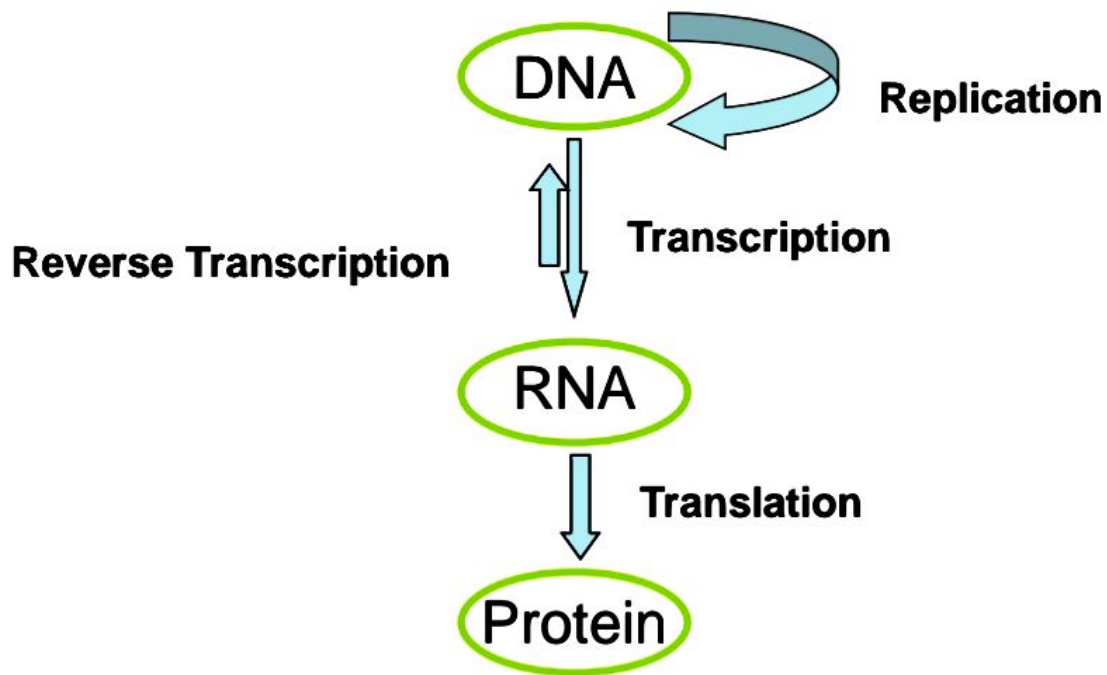
The major intracellular compartments of an animal cell



Relative Volumes Occupied by the Major Intracellular Compartments

INTRACELLULAR COMPARTMENT	PERCENTAGE OF TOTAL CELL VOLUME
Cytosol	54
Mitochondria	22
Rough ER cisternae	9
Smooth ER cisternae plus Golgi cisternae	6
Nucleus	6
Peroxisomes	1
Lysosomes	1
Endosomes	1

The Central Dogma



Protein Biosynthesis

- **Major Requirements are**

Ribosomes

Amino Acids

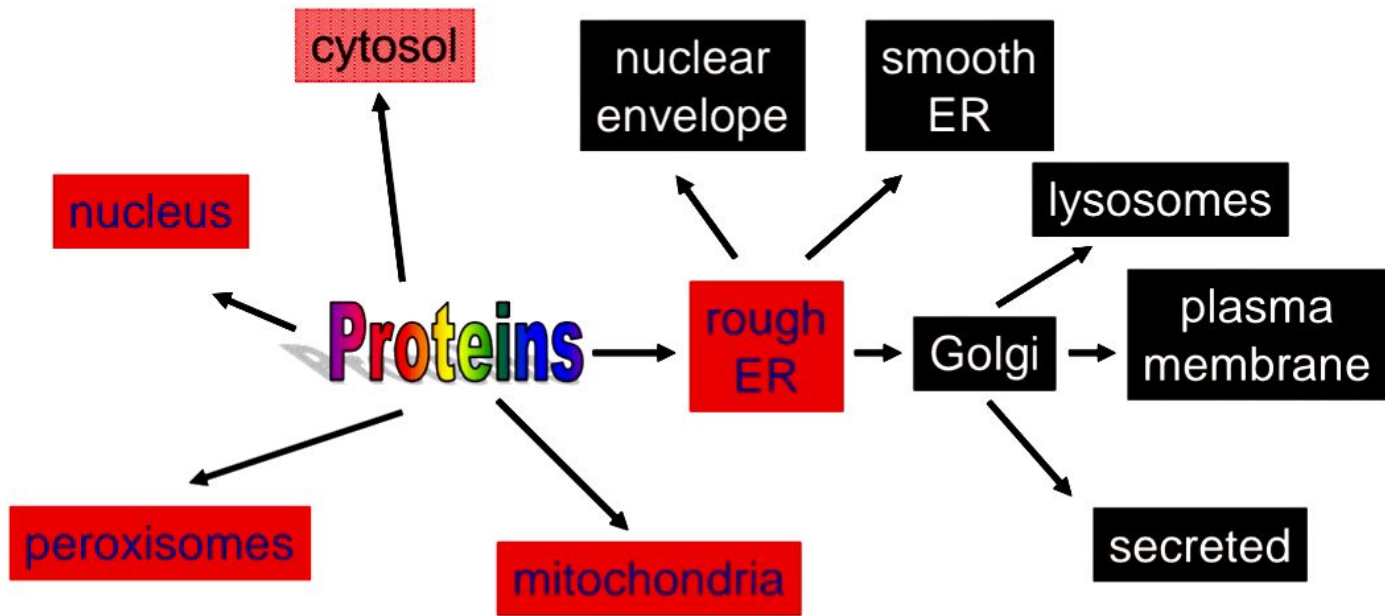
m RNA

t RNA

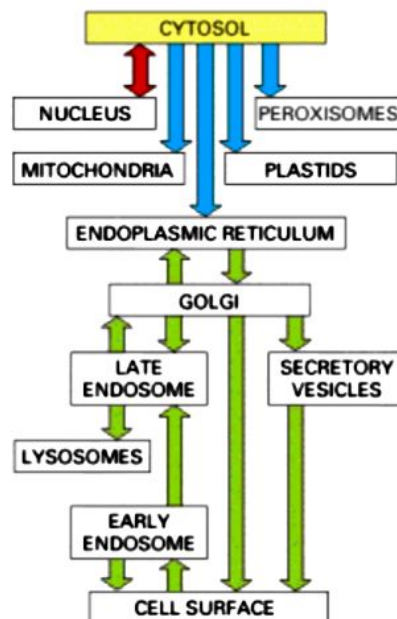
tRNA being the translational adapter is the most important molecule.

Peptide bond formation is thermodynamically unfavourable and therefore amino acids are charged

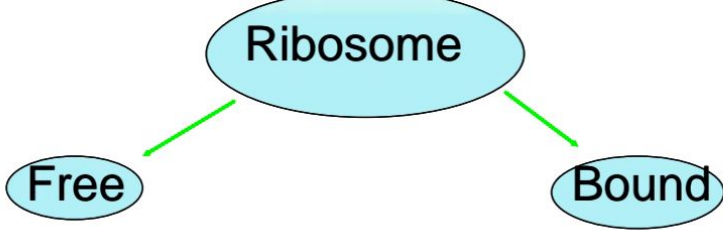
Protein sorting



Protein traffic



KEY: █ = gated transport
█ = transmembrane transport
█ = vesicular transport



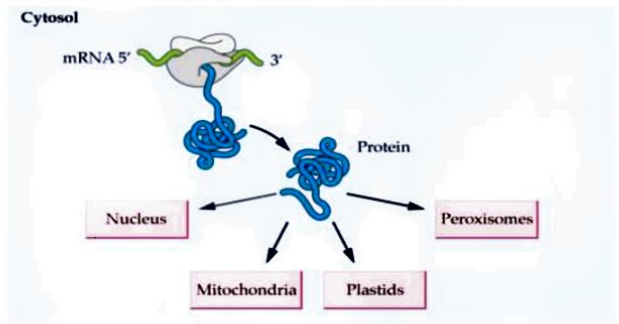
Soluble Proteins

**Lysosomal
Secretory
Plasma Memb.**

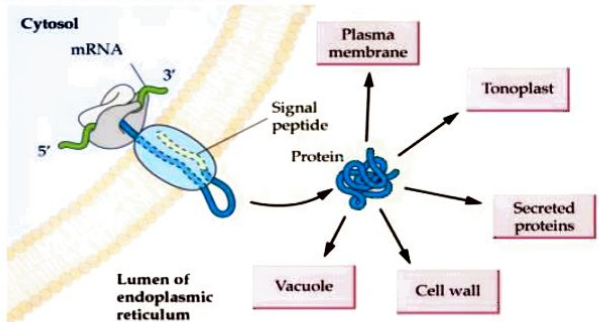
Two types of cytosolic ribosomes: free and membrane-bound.

They synthesize proteins with different destinations.

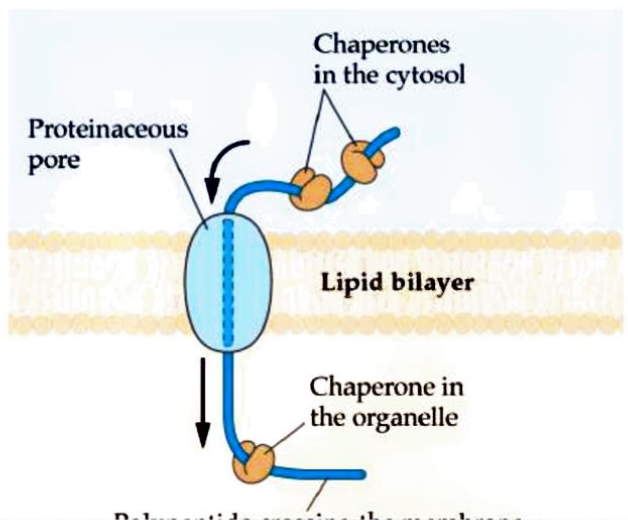
(A) Free ribosomes in cytosol



(B) Membrane-bound ribosomes



2. Chaperonins play roles in membrane transport on both sides of the membrane.



SPECIAL CHROMOSOME: LAMPBRUSH, POLYTENE (SALIVARY GLAND), B-CHROMOSOME

Some tissues of certain organisms contain chromosomes which differ significantly from normal ones in terms of either morphology or function; such chromosomes are referred to as special chromosomes. The following types of chromosomes may be included under this category: (1) Lampbrush chromosomes, (2) Giant chromosome or polytene chromosome and (3) Accessory or B chromosomes.

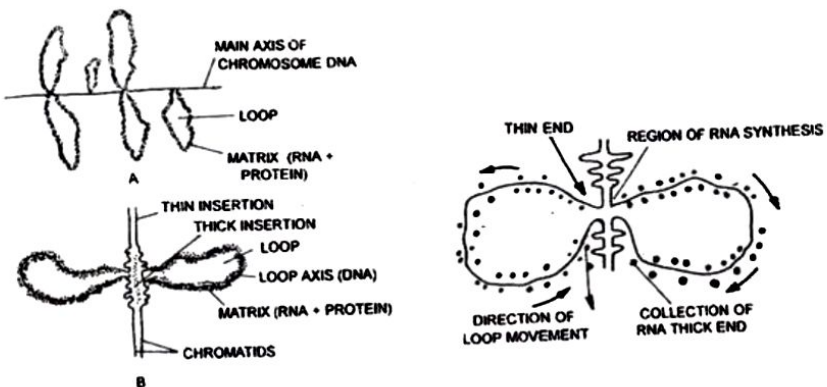
Lampbrush Chromosomes:

Lampbrush chromosomes are found in oocytes of many invertebrates and all vertebrates, except mammals; they have also been reported in human and rodent oocytes. But they have been the most extensively studied in amphibian oocytes. These chromosomes are most distinctly observed during the prolonged diplotene stage of oocytes. During diplotene, the homologous chromosomes begin to separate from each other, remaining in contact only at several points along their length. Each chromosome of a pair has several chromomeres distributed over its length; from each of a majority of the chromomeres generally a pair of lateral loops extends in the opposite directions perpendicular to the main axis of the chromosome. In some cases, more than one pair, even up to 9 pairs of loops may emerge from a single chromomere. These lateral loops give the chromosomes the appearance of a lampbrush which is the reason for their name 'lamp-brush chromosomes.'

These chromosomes are extremely long, in some cases being 800-1000 μ in length. The size of loops may range from an average of 9.5 μ in frog to 200 μ in newt. The pairs of loops are produced due to uncoiling of the two chromatin fibres (hence the two sister chromatids) present in a highly coiled state in the chromosomes; this makes their DNA available for transcription (RNA synthesis). Thus each loop represents one chromatid of a chromosome and is composed of one DNA double helix. One end of each loop is thinner (thin end) than the other end (thick end). There is extensive RNA synthesis at the thin ends of loops, while there is little or no RNA synthesis at the thick end. The chromatin fibre of the chromomere is progressively uncoiled towards the thin end of a loop; the DNA in this region supports active RNA synthesis but later becomes associated with RNA and protein to become markedly thicker. The DNA at the thick end of a loop is progressively withdrawn and reassembled into the chromomere. The number of

1

pairs of loops gradually increases in meiosis till it reaches maximum in diplotene. As meiosis proceeds further, number of loops gradually decreases and the loops ultimately disappear due to disintegration rather than reabsorption back into the chromomere.



Lampbrush chromosome. (A) Gross structure (B) Enlarged view
(C) Synthesis of RNA in a loop of lampbrush chromosome

Mendelian Genetics

- But first, let's introduce a few terms
 - Mendelian factors are now called genes
 - Alleles are different versions of the same gene
 - An individual with two identical alleles is termed homozygous
 - An individual with two different alleles, is termed heterozygous
 - Genotype refers to the specific allelic composition of an individual
 - Phenotype refers to the outward appearance of an individual

Genotype and Phenotype

- Hereditary traits are under the control of genes (Mendel called them factors).
- Genotype is the genetic makeup of an organism, a description of the genes it contains.
- Phenotype is the characteristics that can be observed in an organism.
- Phenotype is determined by interaction of genes and environment.
 - Genes provide potential, but environment determines whether that potential is realized (Figure 2.1).

2.1 MENDEL'S LAWS OF INHERITANCE

- His work, entitled “Experiments on Plant Hybrids” was published in 1866
- Like many great scientific discoveries, it was ignored for 34 years
- In 1900, Mendel’s work was rediscovered by three botanists working independently
 - Hugo de Vries of Holland
 - Carl Correns of Germany
 - Erich von Tschermak of Austria

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Keys to Mendel’s Experiments

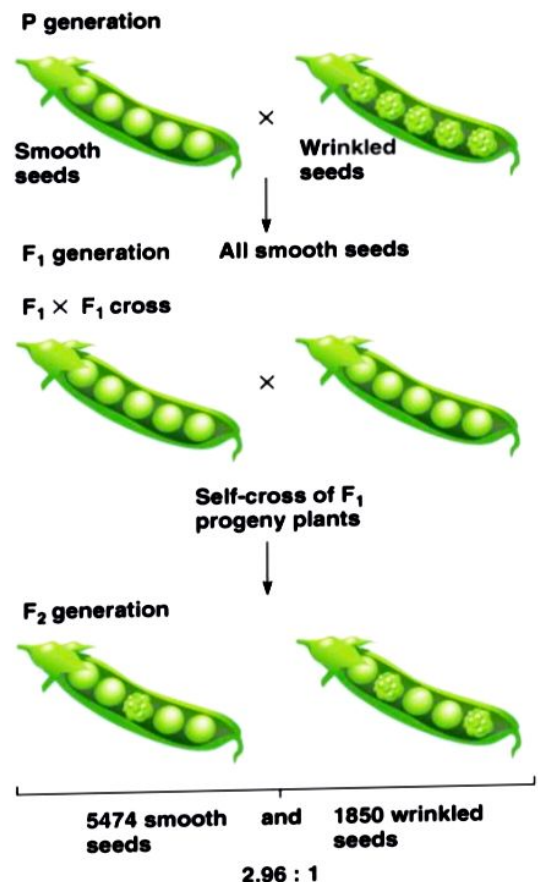
- The garden pea was an ideal organism for study because:
 - Vigorous growth
 - Self fertilization
 - Easy to cross fertilize
 - Produces large number of offspring each generation
- Mendel analyzed traits with discrete alternative forms (one of two options)
 - purple vs. white flowers
 - yellow vs. green peas
 - round vs. wrinkled seeds
 - long vs. short stem length
- Mendel established pure/true breeding lines to conduct his experiments.
 - (traits remain constant from generation to generation)

Monohybrid Crosses and Mendel's Principle of Segregation

- A monohybrid cross involves true-breeding strains that differ in a single trait.
- To determine whether both parents contribute equally to the phenotype of a particular trait in offspring
 - a set of reciprocal crosses is performed.
- In Mendelian genetics, offspring of a monohybrid cross will exactly resemble only one of the parents.
 - This is the principle of uniformity in F₁ (Figure 2.5).
 - Complete dominance

13

Fig. 2.6 The F₂ progeny of the cross shown in Figure 2.5



P



Yellow



Green

Each homozygous parent in the P generation produces only one kind of gamete.

Gametes



F₁



The heterozygous F₁ offspring produces two kinds of gamete.

Gametes



F₂



Self-pollination of the F₁ offspring produces F₂ offspring with a 3:1 ratio of yellow to green seeds.

Ratios

Phenotypes

Genotypes

Genotype ratio

Phenotype ratio

Yellow

YY
Yy

1
2

3

Green

yy

1

1

Meiosis

RrYy

RrYy

SEGREGATION

INDEPENDENT ASSORTMENT

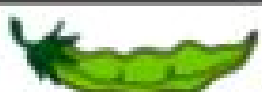
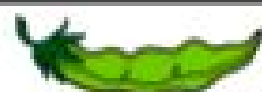
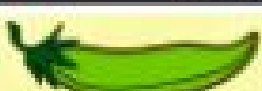
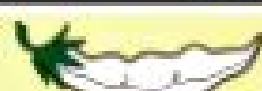
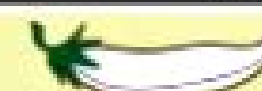
ry

RY

rY

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♀
♂

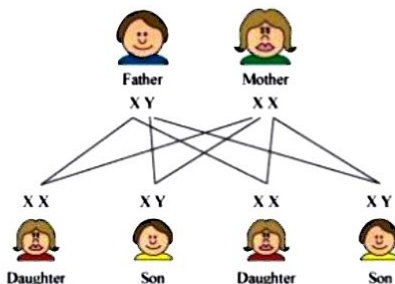
 rryy	 RrYy	 rrYy	 Rryy	ry
 RrYy	 RRYY	 RrYY	 RRYy	RY
 rrYy	 RrYY	 rrYY	 RrYy	rY
 Rryy	 RRYy	 RrYy	 RRyy	Ry
 R	 r	 Y	 y	

Sex Determination

Sex determination, is a system which decides the sexual characteristics of an organism or offspring. It helps to determine whether the organism will be male or a female, which are the two most common sexes. The widely used technique is chromosomal sex determination, in which sex chromosome of male i.e. X or Y chromosome, decides the sex or gender of the offspring. Female carries XX chromosome and male carries XY chromosome. It is also possible to perform genetic tests to eliminate any chromosomal or genetic disorders. Sex determination is the genetic process of determining the sex of the organism. Let us study in more detail about this.

Before we understand how sex determination is done, we need to understand the genetic makeup of a human being. As we all know, humans have 23 pairs or 46 chromosomes. Of these 23 pairs, 22 pairs are known as autosomes whereas 1 pair is known as the sex chromosome. It is this one pair that helps in determining the sex of an individual.

Sex Determination in Humans



Females in humans have 2X chromosomes- 1 each is inherited from either parent and denoted as XX. Males in humans have 1X and 1 Y chromosome, where the X is inherited from the mother and the Y from the father. In a way, we can say that it is the father who determines the sex of the unborn child. This can put to shame a lot of history in which people believed it is the woman who was responsible for not giving birth to a male heir. We can also say that the absence of the Y chromosome makes the individual a female.

At the time of spermatogenesis in males, both types of gametes are produced- one carrying the X chromosome and one carrying the Y. At the time of fertilization, the sex of the resulting zygote will depend on which gamete of the father will fuse with the X of the mother. We can, therefore, say that there is a 50% chance that the child will be a male and 50% that it can be a female.

Types of sex determination

- The XX-XY system as seen in human beings, where, XX is the female and XY is the male. This is also seen in a few insects.

Sex Linkage:

We know that the rules of Mendelian inheritance are not valid for all human traits. Besides the frequency of affected individuals is very different in the two sexes and the appearance of a trait in the offsprings is determined by the fact whether it is passed on by the father or the mother. However, for some of these traits it is now known that though the genes are located on the autosomes yet they have different phenotypic expressions in the two sexes. The traits determined by genes located on the sex chromosomes are called **sex-linked** and their mode of transmission as sex linked inheritance. Thus there are two types of **sex-linked inheritance** depending upon whether the genes are located on X or Y chromosome. It is claimed that in addition to completely X or Y linked genes, there are probably other genes which, by crossing over, are able to change their localization from Y to X chromosome

While considering the details of X and Y linked inheritance it is necessary to explain that a female with XX chromosomes naturally has two alleles at each locus. She may be homozygous or heterozygous for them. But in case of a male, there is only one X chromosome, he is called hemizygous for the single allele of X-linked genes. It may be pointed out that amongst the X and Y chromosomes which are not equal in size, there may exist a small section of Y chromosome which is homologous with the section of the X-chromosome. Thus there could be three possible sites for genes on the sex chromosome.

1. The non-homologous section of the Y-chromosome; such genes are totally Y-linked.
2. The non-homologous section of the X-chromosomes; such genes are totally X-linked.
3. The homologous section of the X and Y-chromosomes; such genes are said to be partially sex-linked as they can pass on from X to Y chromosomes or from Y to X-chromosome or from Y to X-chromosome during crossing over.

- **Sex linkage** describes the sex-specific patterns of inheritance and presentation when a gene mutation (allele) is present on a sex chromosome (allosome) rather than a non-sex chromosome (autosome). In humans, these are termed X-linked recessive, X-linked

dominant and Y-linked. The inheritance and presentation of all three differ depending on the sex of both the parent and the child. This makes them characteristically different from autosomal dominance and recessiveness.

- There are many more X-linked conditions than Y-linked conditions, since humans have several times as many genes on the X chromosome than the Y chromosome. Only females are able to be carriers for X-linked conditions; males will always be affected by any X-linked condition, since they have no second X chromosome with a healthy copy of the gene. As such, X-linked recessive conditions affect males much more commonly than females.
- In X-linked recessive inheritance, a son born to a carrier mother and an unaffected father has a 50% chance of being affected, while a daughter has a 50% chance of being a carrier, however a fraction of carriers may display a milder (or even full) form of the condition due to a phenomenon known as skewed X-inactivation, in which the normal process of inactivating half of the female body's X chromosomes preferably targets a certain parent's X chromosome (the father's in this case). If the father is affected, the son will not be affected, as he does not inherit the father's X chromosome, but the daughter will always be a carrier (and may occasionally present with symptoms due to aforementioned skewed X-inactivation).

Examples

- Aarskog-Scott syndrome
- Adrenoleukodystrophy (ALD)
- Bruton's agammaglobulinemia
- Color blindness
- Complete androgen insensitivity syndrome
- Congenital aqueductal stenosis (hydrocephalus)
- Duchenne muscular dystrophy
- Fabry disease
- Glucose-6-phosphate dehydrogenase deficiency
- Haemophilia A and B
- Hunter syndrome

LINKAGE

- Discovery of linkage
- Meaning of linkage
- Characteristics of linkage
- Genes in linkage
- Theories
- Kinds of linkage
- Linkage group
- Significance

CROSSING OVER

- Discovery of crossing over
- Meaning of crossing over
- Characteristics of crossing over
- Types crossing over
- Mechanisms
- Factors affecting crossing over
- Significance
- Differences between crossing over and linkage

Discovery of Linkage

- The principle of linkage was discovered by English Scientists **William Bateson** and **R.C. Punnet** in 1906 in Sweet Pea (*Lathyrus odoratus*). However, it was put forward as a regular concept by **Morgan** in 1910 from his work on (*Drosophila melanogaster*).



Reginald Crundall Punnett

CHARACTERISTICS OF LINKAGE

- Linkage involves two or more genes which are linked in same chromosomes in a linear fashion.
- Linkage reduces variability.
- It may involve either dominant or recessive alleles(coupling phase) or some dominant and some recessive alleles(repulsion phase).
- It usually involves those genes which are located close to each other.
- The strength of linkage depends on the distance between the linked gene.

Lesser the distance higher the strength of linkage

Genes in Linkage

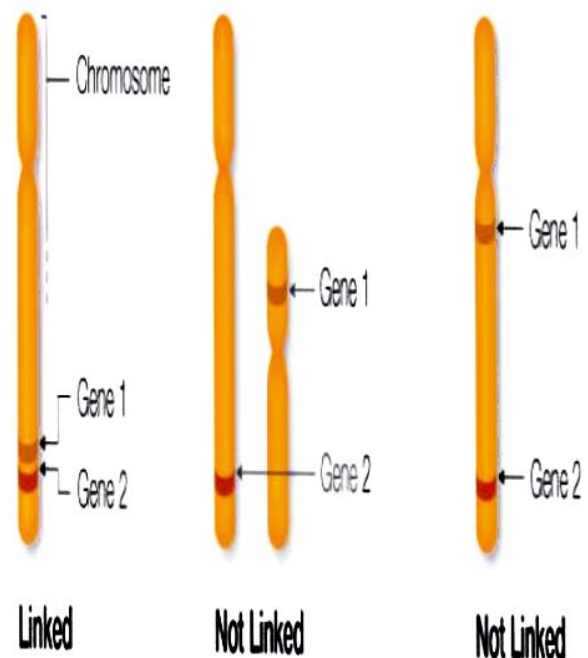
• LINKED GENE :

These genes do not show independent assortment. It occurs in same chromosome.

Dihybrid ratio of linked gene is 3:1

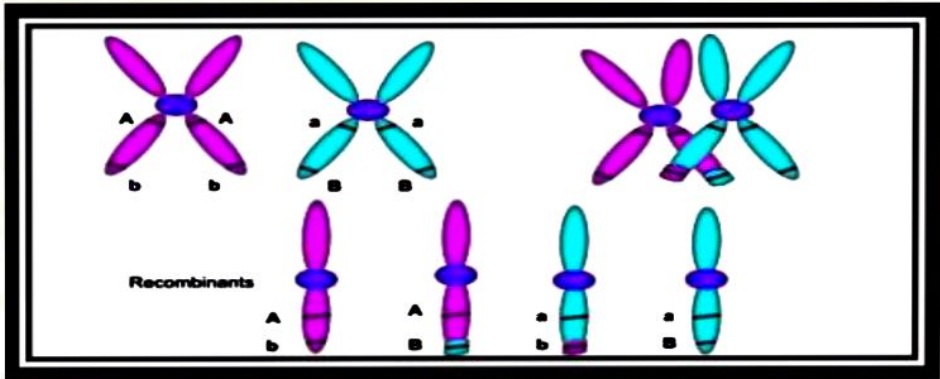
• UNLINKED GENE:

These gene show independent assortment. Dihybrid ratio is 9:3:3:1.



Meaning of Crossing over

- Crossing over or (chromosomal cross over) is the exchange of genetic material between homologous chromosomes that results in recombinant chromosomes.



Characteristics of crossing over

- Crossing over occurs between non-sister chromatids. One chromatid from each of the two homologous chromosomes is involved in crossing over.
- Crossing over leads to re-combinations or new combinations between linked genes.
- The value of crossover or recombinants may vary from 0-50%.
- Crossing over generally yields two recombinant types or crossover types and two parental types or non-crossover types.
- Crossing over generally leads to exchange of equal segments or genes and recombination is always reciprocal.