GENE MAPPING

- Gene mapping is the map/location of genes that are present inside the chromosome.
- Gene mapping is divided into two types:
  - Linkage mapping
  - Physical mapping

Gene Mapping

- Sturtevant proposed that recombination frequencies reflect the distances between genes on a chromosome.
- It is assumed that the chances of crossing over is equal at all points on a chromosome

Gene Mapping

- If so, then **the farther apart two genes are:**
  - (a) the higher the probability that a cross-over will occur between them and therefore
  - (b) the higher the recombination frequency
- Because the greater the distance between two genes the more points there are between them where crossing over can occur

Gene Mapping

- The knowledge of gene linkage can be used to:
  - Determine the order of genes on the chromosome
  - Determine how far apart genes are on the chromosome
  - Construct a map of the chromosome.

Application of Gene Mapping

- Gene mapping is used to locate diseased genotypes
- To locate a particular gene in total genome.
- To screen different types of plants for a desired trait and breed them to get superior products.

Linkage Mapping

- A diagrammatic, graphical representation of relative distances between linked genes of a chromosome is called linkage or genetic map.
- It is based on the use of genetic techniques to construct maps showing the positions of genes and other sequence features on a genome
- Genetic techniques include cross-breeding experiments or in case of humans, the examination of family histories (pedigrees).

Linkage Mapping

- Linkage mapping is the type of mapping which is able to tell us the location of one gene to the other. It tells us whether G1 and G2 are closely related or distantly related. Utilizing linkage map, we can get the distance between two genes.
- Linkage mapping is not satisfactory enough because it can only give us the relative location and relative placement of a gene but it cannot give us the relative presence of a gene inside the chromosome.

Physical Mapping

- Physical map is the type of map which shows the presence of gene inside the chromosome.
- For example,we can tell that G1 is present in chromosome x and G2 is present in chromosome y.
- Physical map can tell us the idea of the exact location of a gene inside the chromosome.
- Physical map is much more specific by giving us the idea of the presence of gene in the chromosome or not.

SEX-LINKED INHERITANCE

- The genes which occur exclusively on the X chromosome are called **X-linked genes**.
- The genes which exclusively occur in Y-chromosome are called **holandric genes**.
- The inheritance of X-linked and holandric genes is called **sex-linked inheritace.**
- In XX-XY type organisms, sex-linked genes can be classified into three types:

SEX-LINKED INHERITANCE

- In XX-XY type organisms, sex-linked genes can be classified into three types:

1. **X-linked**: The X-linked type sex-linked inheritance is performed by those genes which are localized in the non-homologous sections of X-chromosome and that have no corresponding allele in Y-chromosome. The X-linked genes are commonly known as sex-linked genes.
2. **Y-linked:** The Y-linked type sex-linked inheritance is performed by those genes which are localized in the non-homologous section of Y chromosome and that have no alleles in X chromosome. The Y-linked genes are commonly known as holandric genes.

CHARACTERISTICS OF SEX-LINKED (X-LINKED) INHERITANCE

The X-linked genes exhibit following characteristic patterns of inheritance:

1. The genes, whether dominant or recessive, show their effects in the male phenotype.
2. The X-linked recessives can be detected in human pedigrees through the following clues:

CHARACTERISTICS OF SEX-LINKED (X-LINKED) INHERITANCE

(a) The X-linked recessive phenotype is usually found more frequently in the male than in the female.

(b) Usually, none of the offspring of an affected male will be affected, but all his daughters will carry the gene in masked heterozygous condition.

(c) None of the sons of an affected male will inherit the X-linked recessive gene, so not only will they be free of the defective phenotype; but they will not pass the gene along to their offspring.

CHARACTERISTICS OF SEX-LINKED (X-LINKED) INHERITANCE

3. Dominant X-linked genes can be detected in human pedigrees through the following clues:
   (a) It is more frequently found in the female than in the male of the species.
   (b) The affected males pass the condition on to all of their daughters but to none of their sons.
   (c) Females usually pass the condition (defective phenotype) on to one-half of their sons and daughters.
   (d) A X-linked dominant gene fails to be transmitted to any son from a mother which did not exhibit the trait itself.

Examples of Inheritance of X-linked Dominant Genes in Humans

- In humans, X-linked dominant conditions are relatively rare.
- One example is **hypophosphatemia** (vitamin D-resistant rickets).
- Another example includes hereditary **enamel hypoplasia** (hypoplastic amelogenesis imperfecta), in which tooth enamel is abnormally thin so that teeth appear small and wear rapidly down to the gums.

Examples of Inheritance of X-linked Recessive Genes in Humans

- Certain well known examples of X-linked recessive genes in humans are:

1. **Red-green colour blindness** or **daltonism**: Insensitivity to red/green light.
2. **Haemophilia**: More common in men than women. This is known as bleeder's disease. The person which contains the recessive gene for haemophilia lacks in normal clotting substance (thromboplastin) in blood so minor injuries cause continuous bleeding and ultimate death of the person due to haemorrhages.

Examples of Inheritance of X-linked Recessive Genes in Humans

- Two types of X-linked haemophilia have been recognized:
  - **Haemophilia A**: This is characterized by lack of antihaemophilic globulin (factor VIII).
  - **Haemophilia B**: This results from a defect in plasma thromboplastic component (factor IX). This is also called Christmas disease.

Examples of Inheritance of X-linked Recessive Genes in Humans

3. **Duchenne's muscular dystrophy**: This is a progressive, life-shortening disorder characterized by muscle degeneration and weakness; sometimes associated with mental retardation; deficiency of the protein dystrophin.

   Dystrophin is a protein of high molecular weight that is associated with a transmembrane glycoprotein complex of skeletal muscle cells.

Examples of Inheritance of X-linked Recessive Genes in Humans

❑ Some other examples of X-linked recessive traits include:

4. Deficiency of enzyme glucose-6-phosphate dehydrogenase (G6PD deficiency) in erythrocytes causing haemolytic anaemia.
5. Two forms of diabetes insipidus
6. One form of anhidrotic ectodermal dysplasia (absence of sweat gland and teeth)

Examples of Inheritance of X-linked Recessive Genes in Humans

7. Absence of central incisors
8. Certain forms of deafness
9. Spastic parapelagia (i.e., tetanoid)
10. Uncontrollable rolling of the eye balls (nystagmus)
11. A form of cataract
12. Night blindness
13. Juvenile glaucoma (hardening of eye ball)

INHERITANCE OF Y-LINKED GENES

❖ Genes in the non-homologous region of the Y chromosome pass directly from male to male.

❖ In humans, the Y-linked or holandric genes such as **ichthyosis hystrix gravis hypertrichosis** (excessive development of hairs on pinna of ear) are transmitted directly from father to so on.

INHERITANCE OF Y-LINKED GENES

- Certain other holandric genes have been reported in humans, e.g.,
- Histocompatibility antigen
- Spermatogenesis
- Height (stature)
- Slower maturation of indivduals.

INHERITANCE OF X-Y-LINKED GENES

- The genes which occur in homologous section of X and Y chromosome have inheritance like the autosomal genes.
- The X-Y linked genes are **partially** or **incompletely sex-linked** because, sometimes, crossing over may occur in the homologous sections of X and Y chromosomes.

INHERITANCE OF X-Y-LINKED GENES

- In humans, certain diseases are XY-linked. Certain XY-linked genes of man are of
  - Total colour blindness
  - Two skin diseases (*Xeroderma pigmentosum* and *Epipermolysis bullosa*)
  - Retinis pigmentosa, etc.

SEX-INFLUENCED GENES

- Sex-influenced genes are those whose dominance is influenced by the sex of the bearer. Thus, male and female individuals may be similar for a particular trait but give different phenotypic expressions of the same trait.
- For example, in humans, the baldness may occur due to disease, radiation or thyroid defects but in some families, baldness is found to be inherited trait.

SEX-INFLUENCED GENES

- In such inherited baldness, the hairs gradually become thin on head top, leaving ultimately a fringe of hair low on head and commonly known as pattern baldness.
- The gene B for baldness is found to be dominant in males and recessive in females.
- Also, in sheep, the genes for the development of horns is dominant in males and recessive in females.

SEX-LIMITED GENES

- Sex-limited genes are autosomal genes whose phenotypic expression is determined by the presence or absence of one of the sex hormones. Their phenotypic effect is limited to one sex or the other.
- Sex-limited genes are mainly responsible for secondary sex characters in cattle, humans and fowl.

SEX-LIMITED GENES

- For example, the bulls have genes for milk production which they transmit to their daughters, but they or their sons are unable to express this trait. The production of milk is therefore limited to variable expression only in the female sex.
- Beard development in human beings is a sex-limited trait as men normally have beards, whereas women do not.

SEX-LIMITED GENES

- Likewise, the genes for male voice, body hair and physique are autosomal in human beings, but they are expressed only in the presence of androgens which are absent in females.
- In chicken, the recessive gene (h) for cock feathering is male sex-limited (i.e., it is penetrant only in male environment).

MULTIPLE CHOICE QUESTIONS

1. In *Drosophila*, the sex-linked inheritance was observed by

   (a) T.H. Morgan (b) Baldeyer (c) Kornberg (d) Calvin

2. The sex chromosomes (X and Y) are of unequal size, shape or staining quality. Hence, it is said to be

   (a) autosomes (b) heteromorphic

   (c) allelomorphic (d) metamorphic

MULTIPLE CHOICE QUESTIONS

3. Sex-linked characters are

   (a) dominant (b) recessive (c) lethal

   (d) not inherited

4. Red green colour blindness appears due to

   (a) excessive drinking of alcohol

   (b) inheritance through X chromosome

   (c) over activity of adrenal

   (d) vitamin A deficiency

MULTIPLE CHOICE QUESTIONS

5. Criss-cross inheritance pattern is more prevalent and related to

   (a) X-linked genes (b) Y-linked genes

   (c) XY-linked genes (d) XX-linked genes

6. Which of the following diseases belong to the same category as colour blindness in man?

   (a) night blindness (b) presbyopia

   (c) diabetes incipidus (d) haemophilia

CELL DIVISION- MITOSIS AND MEIOSIS

- The asexual cell division in eukaryotes is called **mitosis** (M).
- The sexual cell division in eukaryotes is called **meiosis** (Mei) and occur in specialized cells, called the meiocytes, which divides twice, resulting in four haploid cells called a **tetrad.**

MITOSIS & MEIOSIS

- Mitosis is a kind of cell division that occurs in the somatic (body) cells of all plants and animals during growth, development, repair and asexual reproduction.
- Meiosis, on the other hand, takes place in the germinal cells (gonads) of organisms during gametogenesis, i.e., formation of gametes- sperms and ova/eggs.

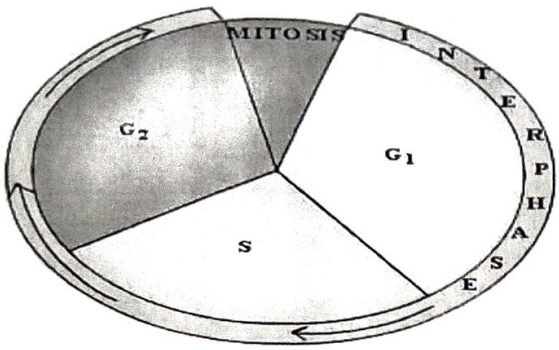
MITOSIS & MEIOSIS

- Each of these cell divisions includes two events:
  - Nuclear division or karyokinesis, which is followed by the
  - Division of the Cytoplasm or Cytokinesis.

THE CELL CYCLE

- Actively dividing eukaryote cells pass through a series of stages known collectively as the **cell cycle, it comprises of :**
  - two gap phases (G1 and G2);
  - an S (for synthesis) phase, in which the genetic material is duplicated; and
  - an M phase, in which mitosis partitions the genetic material and the cell divides.

The Cell Cycle



The Cell Cycle- I phase

- Interphase (I-Phase): This is the period between mitotic divisions - that is, G1, S and G2.
- It is a period of synthesis (of DNA, rRna, tRna, mRna and proteins such as DNA polymerase,histones, etc.) and growth.
- The cell produces many materials(including cell organelles) required for its own growth and for carrying out all its functions.

The Cell Cycle-G1 phase

- G1 phase is a resting phase and it is also the longest phase. It is called the gap phase since no DNA synthesis takes place during this phase.
- G1 phase is also called first growth phase, since it involves synthesis of RNA (transcription), proteins and membranes, which leads to the growth of nucleus and cytoplasm of each daughter cell towards their mature size.
- G1 is a post-mitotic gap phase which takes place at the end of one cell division.

The Cell Cycle- S phase

- S phase or synthetic phase is the stage when **DNA synthesis** (chromosome replication) occurs. The chromosomes consist of two identical strands once replication is completed. Each of these strands is called a **chromatid.**
- During mitosis the chromatids will separate and each chromatid will become **a separate chromosome.**

The Cell Cycle- G2 phase

- G2 phase is a pre-mitotic gap phase, second gap phase or second growth phase. It is also called resting phase or second interval because no DNA replication takes place.
- During G2 phase, synthesis of RNAs and proteins continue; mitochondria and chloroplasts divide; stores of energy increase and mitotic spindle begins to form.

The Cell Cycle-M phase

- Mitotic (M) phase is the period of nuclear division (karyokinesis), which is followed by cell divison (cytokinesis).
- Cytokinesis involves equal distribution of organelles and cytoplasm into each daughter cell.

MITOSIS

- Mitosis is a process of nuclear division (karyokinesis) in whichreplicated DNA molecules of each chromosomes are faithfully partitioned into two nuclei.
- Karyokinesis of mitosis is usually followed by cytokinesis, a process by which dividing cell splits into two, partitioning the cytoplasm into two packages.

MITOSIS

- The two daughter cells resulting from mitosis and cytokinesis possess a genetic content identical to each other and to the mother cell from which they arose.
- Mitosis maintains the chromosome number and generates new cells for growth and maintenance of an organism (e.g., blood cells, skin cells, etc.).
- Mitosis can take place in either haploid and diploid cells. Haploid mitotic cells are found in fungi, plant gametophytes and a few animals(such as male bees known as drones).

Stages of Mitosis

- Mitosis is generally divided into five stages:
  - Prophase
  - Prometaphase
  - Metaphase
  - Anaphase
  - Telophase.

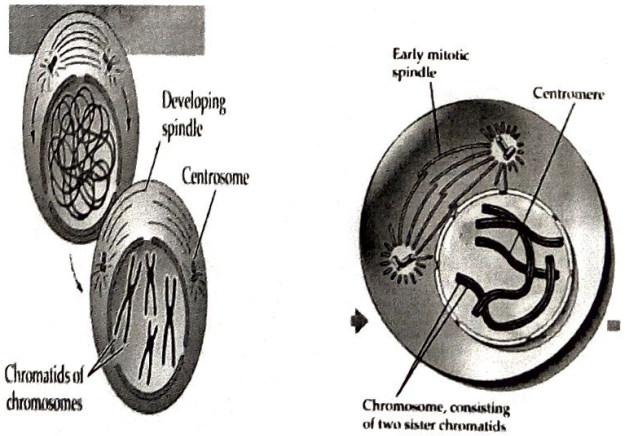
Prophase

- The nuclear membrane breaks down to form a number of small vesicles and the nucleolus disintegrates.
- A structure known as the centrosome duplicates itself to form two daughter centrosomes that migrate to opposite ends of the cell.

Prophase

- The centrosomes organise the production of microtubules that form the spindle fibres that constitute the mitotic spindle.
- The chromosomes condense into compact structures.
- Each replicated chromosome can now be seen to consist of two identical chromatids (or sister chromatids) held together by a structure known as the centromere.

Prophase



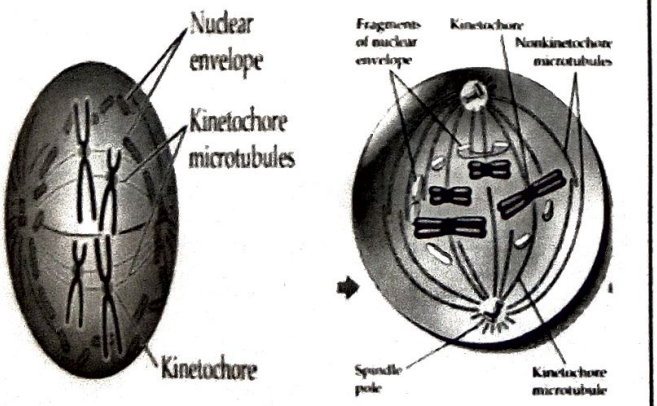
Prometaphase

- The chromosomes, led by their centromeres, migrate to the equatorial plane in the midline of cell - at right-angles to the axis formed by the centrosomes.
- The spindle fibres bind to a structure associated with the centromere of each chromosome called a kinetochore.

Prometaphase

- Individual spindle fibres bind to a kinetochore structure on each side of the centromere.
- The chromosomes continue to condense.
- Nuclear envelope disappears
- Nucleolus disappears

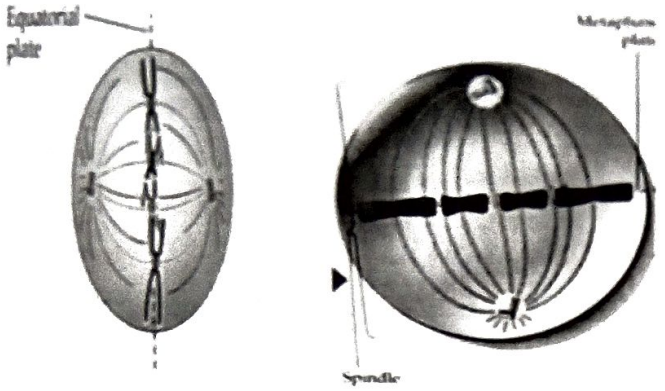
Prometaphase



Metaphase

- The chromosomes move to the equatorial plane of the cell, where one sister centromere becomes attached to a spindle fiber from one pole; the other sister centromere, to the other pole.

Metaphase



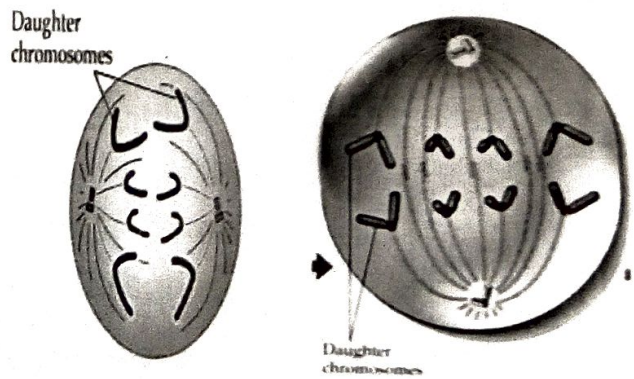
Anaphase

- This is the shortest stage of mitosis.
- The centromeres divide, and the sister chromatids of each chromosome are pulled apart - or 'disjoin' - and move to the opposite ends of the cell, pulled by spindle fibres attached to the kinetochore regions.

Anaphase

- The separated sister chromatids are now referred to as **daughter chromosomes.**
- (It is the alignment and separation in metaphase and anaphase that is important in ensuring that each daughter cell receives a copy of every chromosome.)

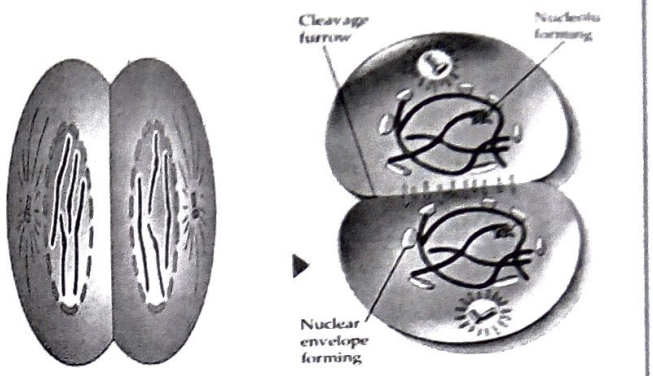
Anaphase



Telophase and Cytokinesis

- The final stage of mitosis, and a reversal of many of the processes observed during prophase.
- The nuclear membrane reforms around the chromosomes grouped at either pole of the cell,
- The nucleolus reforms
- The chromosomes uncoil and become diffuse, and
- The spindle fibres disappear.
- A cleavage furrow divides the cytoplasm in two = **cytokinesis**

Telophase



MEIOSIS

- Meiosis consists of two nuclear divisions, distinguished as meiosis I and meiosis II. The events of meiosis I are quite different from those of meiosis II, and the events of both differ from those of mitosis.
- Before undergoing the meiosis I, each meiocyte remains in the interphase during which the genetic materials are duplicated due to active DNA replication.

MEIOSIS

- Each meiotic division is formally divided into:
  - Prophase
  - Metaphase
  - Anaphase, and
  - Telophase.

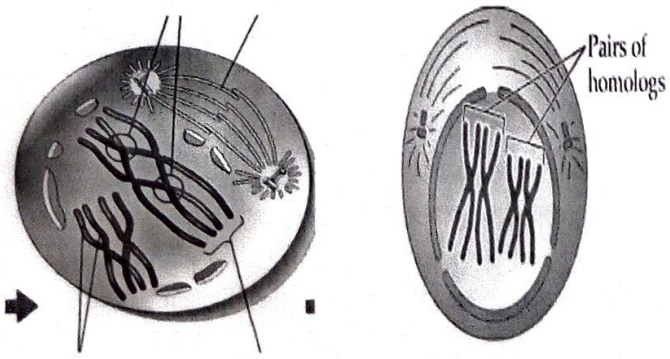
PROPHASE I.

- ❑ The homologous chromosomes pair and exchange DNA to form recombinant chromosomes.
- ❑ Prophase I is divided into five phases:
  - ➢ **Leptotene**: chromosomes start to condense.
  - ➢ **Zygotene**: homologous chromosomes become closely associated (synapsis) to form pairs of chromosomes (bivalents) consisting of four chromatids (tetrads).

PROPHASE I.

- **Pachytene**: crossing over between pairs of homologous chromosomes to form chiasmata (sing. chiasma).
- **Diplotene**: homologous chromosomes start to separate but remain attached by chiasmata.
- **Diakinesis**: homologous chromosomes continue to separate, and chiasmata move to the ends of the chromosomes.

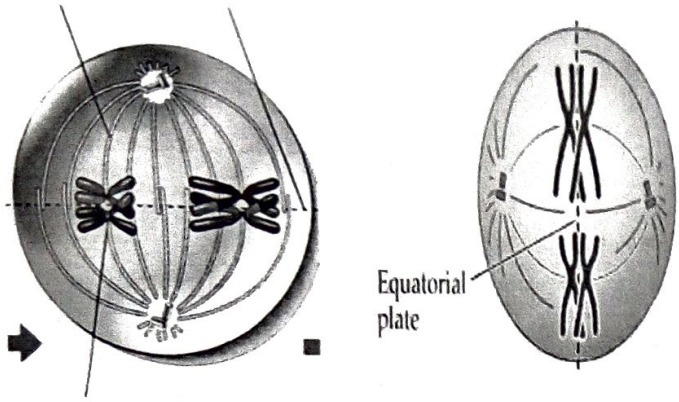
PROPHASE I.



METAPHASE I.

- ❑ The nuclear membrane and nucleoli disappear, and each pair of homologs takes up a position in the equatorial plane.
- ❑ The sister centromeres do not appear to have divided, so they act as one. This apparent lack of division represents a major difference from mitosis.
- ❑ The two nonsister centromeres attach to spindle fibers from opposite poles.

METAPHASE I.

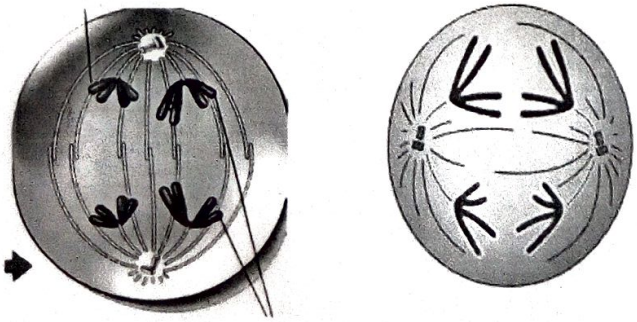


ANAPHASE I.

- Sister chromatids remain attached
- But homologous chromosomes move apart

to opposite poles

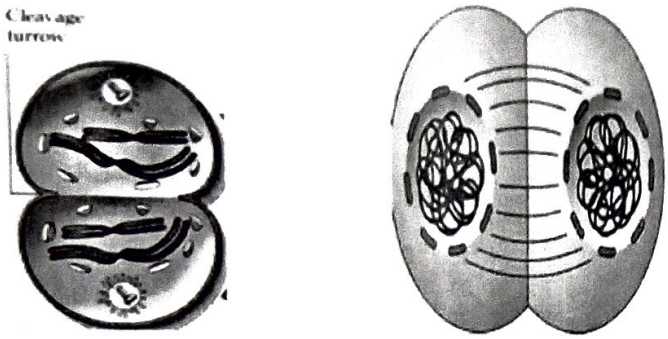
ANAPHASE I.



TELOPHASE I.

- In many organisms, this stage do not exist, no nuclear membrane re-forms, and the cells proceed directly to meiosis II.
- In other organisms, telophase I and the interkinesis are brief in duration; the chromosomes elongate and become diffuse, and the nuclear membrane re-forms.
- In any case, there is never DNA synthesis at this time, and the genetic state of the chromosomes does not change.

TELOPHASE I.



Cytokinesis

- ❑ This is the final cellular division to form two new cells, followed by Meiosis II.
- ❑ Meiosis I is a reduction division: the original diploid cell had two copies of each chromosome; the newly formed haploid cells have one copy of each chromosome

Meiosis II

- Meiosis II separates each chromosome into two chromatids.

Prophase II

- ❑ Chromosomes begin to condense
- ❑ Nuclear membrane dissolves
- ❑ Spindle fibres form

Metaphase II

- Spindle fibres attach to chromosome
- Chromosomes line up in the center of the cell

Anaphase II

- Centromeres divide and sister chromatids move to opposite ends of the cell as the spindle fibres shorten

Telophase II

- Chromosomes reach the opposite ends of the cell
- Nuclear membranes form.

MEIOSIS II