

UNIT-3 (Genetics)

The seven Characters Mendel studied.

Trait	Shape of seeds	Colour of seeds	Colour of pods	Shape of pods	Plant height	Position of flower	Flower colour
Dominant trait	Round	Yellow	Green	Full	Tall	At leaf junctions	Purple
Recessive trait	Wrinkled	Green	Yellow	Flat, constricted	Short	At tips of branches	White

Genetic crosses:

- ⇒ To cross two different pea plants, Mendel used an artist's brush
- ⇒ He transferred pollen from a true breeding white flower to the carpel of a true breeding purple flower.

Mendel's terminology:

1. True breeding: When the plants self pollinate, all their offspring are of the same variety.
2. Hybridisation: Mating, or crossing, of two varieties.
3. Monohybrid cross: A cross b/w two plants that breed true for different versions of a single trait.

Tracking characteristics:

- ⇒ Mendel tracked heritable characters for 3 generations
- ⇒ When F₁ hybrids were allowed to self pollinate a 3:1 ratio of the 2 varieties occurred. In the F₂ generation.

P/Generation
(true breeding parents)

What is Genetics:

All body cells contain "Blue prints" with instructions as to how an animal will look or act etc

One gene comes from each parent (pairs)

Genes are divided into sections (Chromosomes) that carry genes*

Genotypes:

The genotype refers to the entire set of genes in a cell, an organism, or an individual. A gene for a particular character or trait may exist in two forms, one is dominant (E) & the other is recessive (e).

Examples:

There are three basic genotypes

for a particular character : A

AA = homozygous dominant

Aa = heterozygous

aa = homozygous ~~to~~ recessive

A	AA	Aa
	AA	Aa

Phenotypes:

Phenotype is the physical appearance or other characteristic of an organism ~~are~~ as a result of the interaction of its genotype and the environment.

Some eg. would be

- (i) Size
- (ii) Shape
- (iii) Colour.

Dominant And Recessive Genes

Dominant genes: One gene overshadows the other.

Recessive Genes: The gene that is overshadowed by a dominant gene.

The Punnet Square

It is a square grid used in genetics to calculate the frequencies of the different genotypes and phenotypes among the offspring of a cross

Mendel's Laws

1. Law of Dominance: If the two alleles at a locus differ, then one, the dominant allele, determines the organism's appearance; the other, the recessive allele, has no noticeable effect on the organism's appearance.
2. Law of Segregation: The two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes.
3. Law of Independent Assortment: each pair of alleles segregates independently of other pairs of alleles during gamete formation.

Blood Groups

Blood groups, also called blood types, can be defined as the grouping of blood and are based on the presence or absence of genetically defined antigenic substances on the surface of red blood cells (RBC).

The antigenic substances may either be carbohydrates, glycolipids, proteins, or glycoproteins, depending on the blood type system.

* Karl Landsteiner, an Austrian scientist discovered the ABO blood group system in the year 1900. For this discovery, he was awarded the Nobel Prize.

Genetics of Blood Group.

ABO blood group antigens present on RBC.

	Group A	Group B	Group AB	Group O
Red Blood cell type				
Antibodies in Plasma			NONE	
Antigens in Red Blood Cell	A antigen	B antigen	A & B antigens	NONE D-antigen

Inheritance of the ABO Blood system in Humans.

I^A	I^B	i	
I^A	$I^A I^A$	$I^A I^B$	$I^A i$
I^B	$I^B I^A$	$I^B I^B$	$I^B i$
i	I^A	I^B	O

FACTS:

1. A newborn baby has only one cup of blood in the whole body, whereas, the healthy adult has about 1.3 to 1.6 gallons or 4.0 to 5.0 liters of blood circulating inside their body.
2. Blood type influences one's personality, which can also affect your fertility and belly fat. Women with blood type O are more likely to deal with fertility issues.
3. The Red Blood cells that do not contain either A or B antigens on their surface are normally found in the person with blood type O.
4. Anybody with good health weight and around the age of 17 years old can donate blood every 3 to 4 months.
5. More than 4.5 million lives are saved every year by blood transfusion.
6. O blood type is called to be the "universal donor" because O^+ is the most frequently occurring ~~blood~~ blood group in the ABO type. About 37 to 40%.

of the entire population are found with this type of blood type. O⁻ is the rare blood group as it is found in 6-7% of the total population.

7. Transfusion with a blood group different from yours can lead to complications.
8. In ABO blood type, a transfusion of AB blood group can be given to a person who has blood type A, B and AB.
9. AB blood type is known to be the "universal recipient" because AB⁺ people can accept blood from any other blood type. AB is the least common blood type, which is less than 1% of the population. An individual with AB-blood type can receive blood from all three -ve blood types.
10. A⁺ is the third most frequently occurring blood type in the ABO system. Thirty of every hundred people have A⁺ blood type. A⁻ is the rare blood type, which is less than 10-15% of the population has this blood type.
11. Both B⁺ & B⁻ are rare blood types and less than 10% of the population has this blood type.
12. Like hair colour, texture, and eye colour, blood type is also inherited or passed genetically from our parents. Therefore, our blood group is based on the blood types of either mother or father.

Diabetes

Diabetes Mellitus

It is a metabolic disorder characterized by hyperglycemia due to an absolute or relative lack of insulin or to a cellular resistance to insulin.

Major Classifications:

1. Type 1 Diabetes
2. Type 2 Diabetes

Difference b/w Type 1 & 2 Diabetes

Type 1: It is known as insulin-dependent and is caused by deficient insulin production & requires daily injection of insulin.

Type 2: It is formally called non-insulin-dependent diabetes from the body's ineffective use of insulin and is largely the result of excess body weight and physical inactivity.

• Type 2 Diabetes Mellitus

Was previously called non-insulin-dependent diabetes mellitus (NIDDM) or adult-onset diabetes.

- ⇒ In adults, type 2 diabetes accounts for about 90% to 95% of all diagnosed cases of diabetes.
- ⇒ Usually begins as insulin resistance, a disorder in which the cells do not use insulin properly.
- ⇒ As the need for insulin rises, the pancreas gradually loses its ability to produce insulin.
- ⇒ It is associated with older age, obesity, family history

age,民族, history of gestational diabetes, impaired glucose metabolism, physical inactivity and race/ethnicity.

Genetic Disorders

A genetic disorder is an illness caused by one or more abnormalities in the genome, especially a condition that is present from birth (congenital).

Most genetic disorders are quite rare and affect one person in every several thousands or millions.

Genetic disorders may or may not be heritable, i.e. passed down from the parent's genes.

Classification:

- (i) Single gene disorder
- (ii) Chromosomal genetic disorder
- (iii) multifactorial genetic disorder.

(i) Single Gene Disorders:

A mutation causes the protein product to be altered or missing

(mutation = sudden change)

(ii) Chromosomal Genetic disorder:

Entire chromosomes, or large segments of them are missing, duplicated, or altered.

(iii) Multifactorial genetic disorder:

Result from mutations in multiple genes coupled with environmental causes.

Single Gene Disorder

These disorders involve mutations in the DNA sequences of single genes.

Over 4000 human diseases caused by single gene defects.

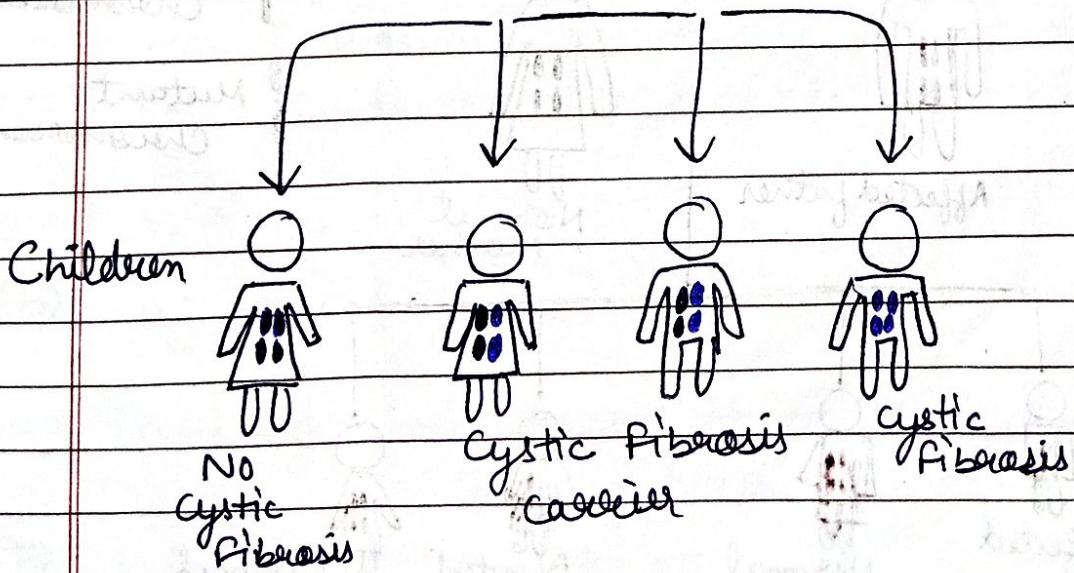
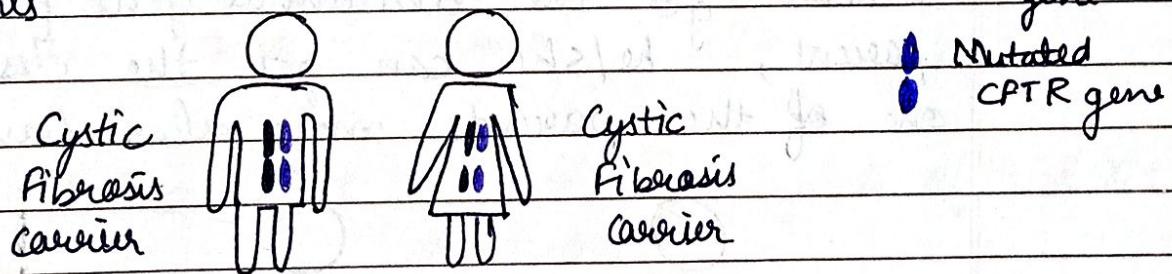
Some examples: Cystic fibrosis, Huntington's disease, sickle cell etc.

Cystic Fibrosis

(large mucus production followed by salt)

- ⇒ Cystic fibrosis is a genetic disorder that affects the respiratory and digestive systems.
- ⇒ People with cystic fibrosis inherit a defective gene on chromosome 7 called CFTR (cystic Fibrosis transmembrane conductance regulator).
- ⇒ The protein produced by this gene normally helps salt (sodium chloride) move in & out of cells.

Parents



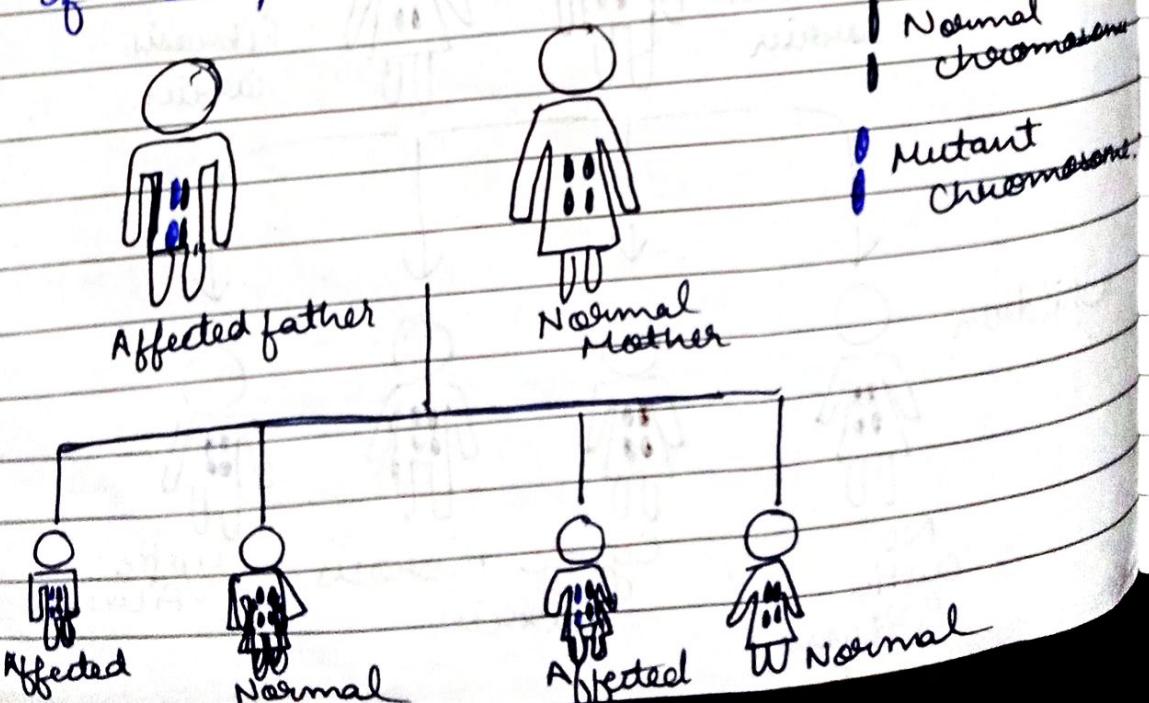
The three common types of single gene disorders are called :

- (i) Autosomal dominant
- (ii) Autosomal recessive
- (iii) X-linked

- ⇒ An autosome is a chromosome that is not an allosome (i.e. not a sex chromosome).
- ⇒ Autosomes appear in pairs.
- ⇒ Humans have a diploid genome that usually contains 22 pairs of autosomes and one allosome pair (46 chromosomes total).
- ⇒ Disorders related to Autosome are autosomal disorders.

(i) Autosomal Dominant :

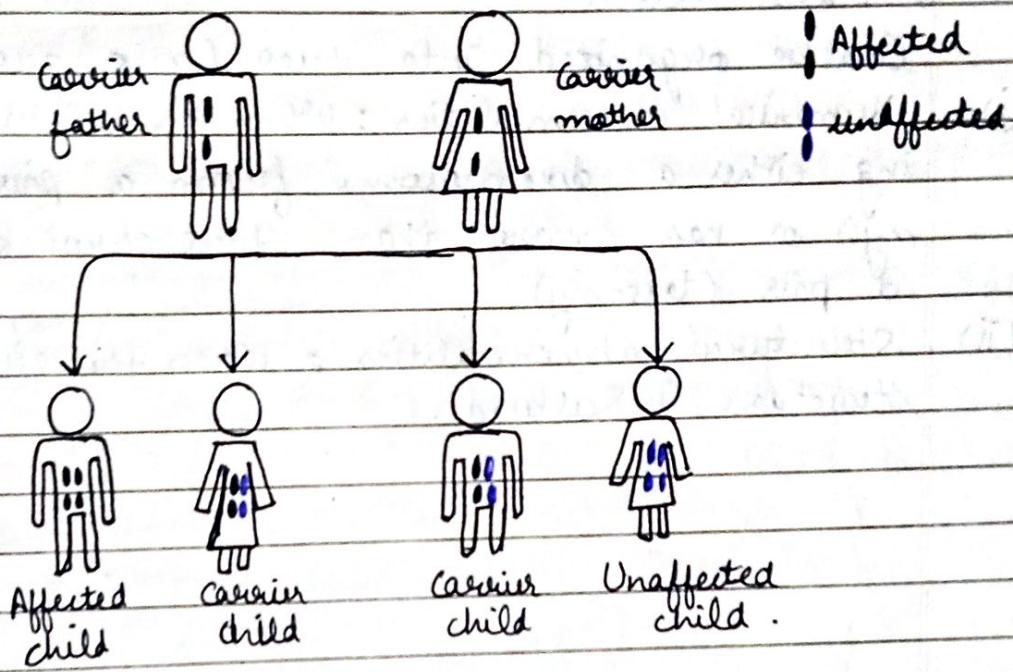
Autosomal Dominant is one of many ways that a trait or disorder can be passed through families. In an autosomal dominant disease, if someone gets the abnormal gene from only one parent, he/she can get the disease. Often one of the parents may also have the disease.



orders

(ii) Autosomal Recessive :

- ⇒ Two mutated copies of the genes are present in a person has an autosomal recessive disorder.
- ⇒ An affected person usually has unaffected parents who each carry a single copy of the mutated gene.
- ⇒ Typically not seen in every generation of an affected family.



(iii)(a) X-linked Dominant : mutations on genes of the X-chromosome.

- ⇒ Men & women are equally likely to get it
- ⇒ Rett syndrome (progressive brain disorder)
- ⇒ Fragile X

(b) X-linked Recessive : mutations on the genes of the X chromosome.

- ⇒ Males are more likely to get these than women
- ⇒ Duchene Muscular Dystrophy
- ⇒ Hemophilia

- c) Y-linked :- mutations in genes on the Y chromosome
- ⇒ All males will have these disorders
- ⇒ Rare since the Y chromosome is small.

Chromosomal Genetic Disorder

In these disorders, entire chromosomes, or large segments of them, are missing, duplicated, or otherwise altered.

Can be organised into two basic groups:

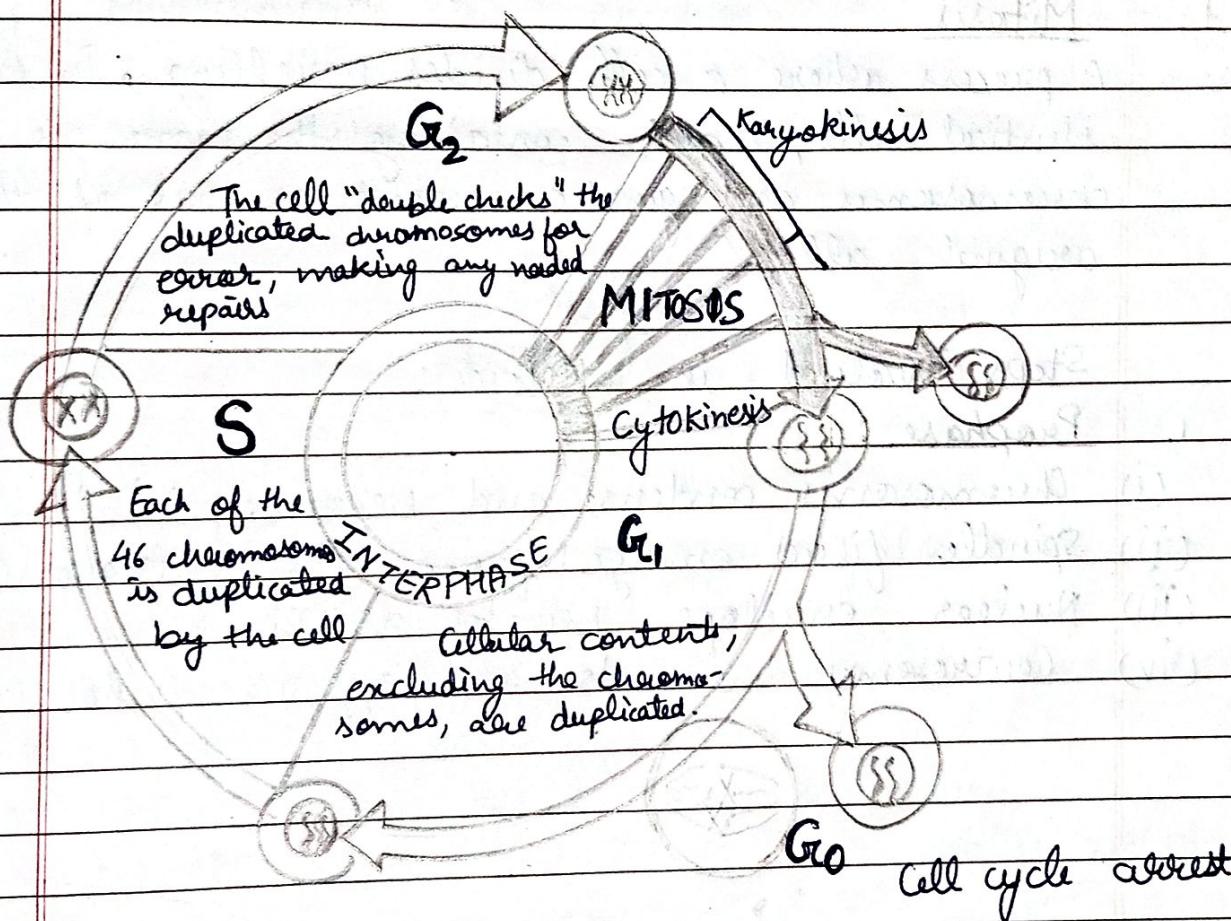
- (i) Numerical abnormalities : When an individual is missing either a chromosome from a pair (monosomy) or has more than two chromosomes of a pair (trisomy).
- (ii) Structural abnormalities : When the chromosome's structure is altered.

Cell Division

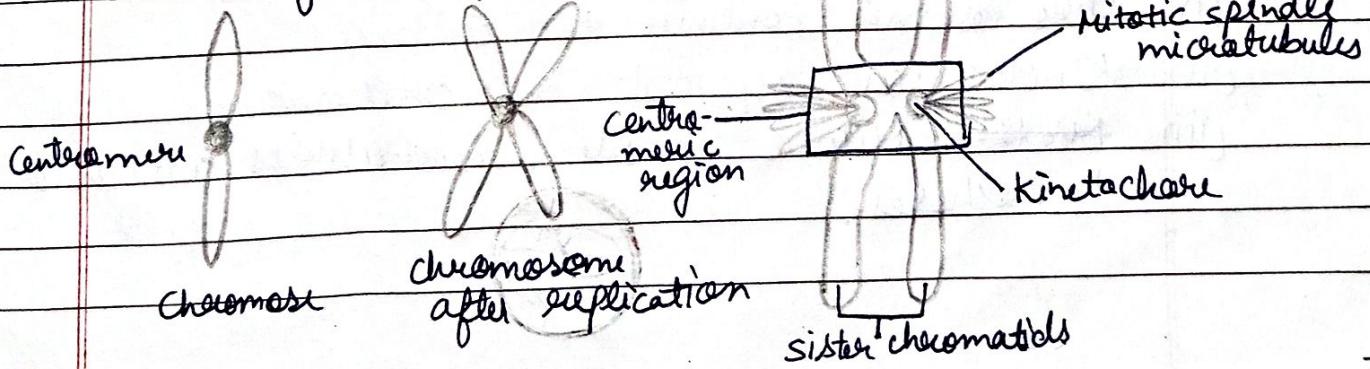
All cells arise from other cells by cell division. When cells grow to a certain size they must either divide or die.

If a cell continued to grow without dividing, the surface area of the cell would become too small to hold the cell's contents.

Cell cycle:



Structure of chromosome:



Cytokinesis: In the cytokinesis process the cytoplasm of single eukaryotic cell divides to form two daughter cells.

Types of Cell Division

1. Mitosis
2. Meiosis

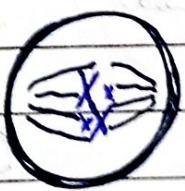
1. Mitosis

A process where a cell divides resulting in two identical cells, each containing the same no. of chromosomes and genetic content as that of the original cell.

Steps involved in Mitosis:

1. Prophase:

- (i) Chromosomes condense and become visible
- (ii) Spindle fibres emerge from the centrosomes
- (iii) Nuclear envelope breaks down.
- (iv) Centrosomes move toward opposite poles.



2. Metaphase:

- (i) Chromosomes continue to condense
- (ii) Kinetochores appear at the centromeres.
- (iii) ~~Anaphase~~ Mitotic spindle microtubules attach to kinetochores.



3. Metaphase :

- (i) Chromosomes are lined up at the metaphase plate.
- (ii) Each sister chromatid is attached to a spindle fiber originating from opposite poles.



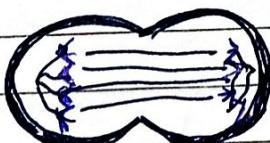
4. Anaphase :

- (i) Centromeres split in two.
- (ii) Sister chromatids (now called chromosomes) are pulled toward opposite poles.
- (iii) Spindle fibers begin to elongate the cell.



5. Telophase :

- (i) Chromosomes arrive at opposite poles and begin to decondense.
- (ii) Nuclear envelope material ~~surrounds~~ surrounds each set of chromosomes.
- (iii) The mitotic spindle breaks down.
- (iv) Spindle fibers continue to push poles apart.



All these steps of mitosis comes under the process karyokinesis.
The next step involved in mitosis is known as cytokinesis.

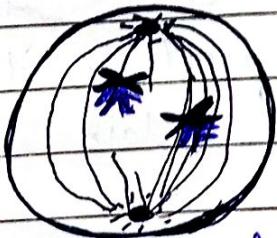
Cytokinesis:

- ⇒ Animal cells: a cleavage furrow separates the daughter cells.
- ⇒ Plant cells: a cell plate, the precursor to a new cell wall, separates the daughter cells



2. Meiosis

Meiosis is the process of division of a cell in two stages that results in four cells, each with half the chromosomes of the original cell.



Meiosis I

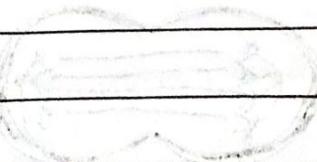
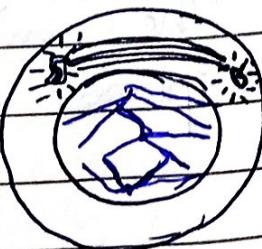
1. Prophase I.

The prophase stage of meiosis I is relatively long and can be subdivided into five stages:

(a) Leptotene (thin thread):

⇒ The chromosomes become visible, shorten and thicken.

⇒ The size of the nucleus increases and homologous chromosomes start getting closer to each other.



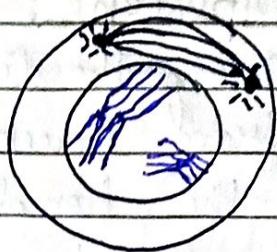
(b) Zygotene

⇒ The zygotene stage, also known as zygotenna, from Greek words meaning "paired threads."

⇒ Zygotene, occurs as the chromosomes approximate line up with each other into homologous chromosome pairs.

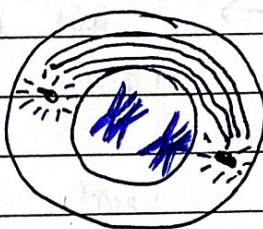


The combined homologous chromosomes are said to be bivalent.



(c) Pachytene

- ⇒ In pachynema, the homologous chromosomes become much more closely associated. This process is known as synapsis.
- ⇒ The synapsed homologous pair of chromosomes is called a tetrad, because it consists of four chromatids.
- ⇒ It can't be observed until the next stage, but the synapsed chromosomes may undergo crossing over in pachynema.
- ⇒ The chromosomes continue to condense.



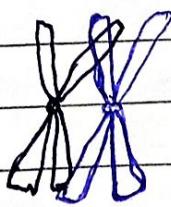
What exactly is synapsis and crossing over?

Synapsis is the connecting of homologous chromosomes to form a tetrad.

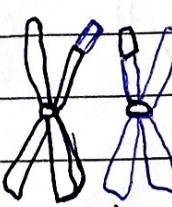
Crossing over is the sharing of genetic material between two non-sister chromatids in a homologous pair.



Homologous pair



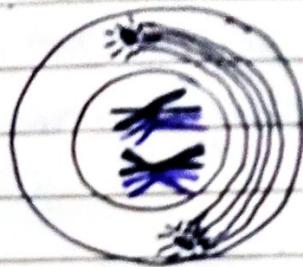
Synapsis



Crossing over

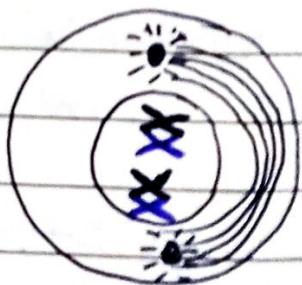
(d) Diplotene:

- ⇒ During the diplotene stage, also known as diplaconia, from Greek words meaning "two threads."
- ⇒ the homologous ~~chromatids~~ chromosomes separate from one another a little.
- ⇒ the chromosomes themselves uncoil a bit, after some transcription of DNA.



(e) Diakinesis

- ⇒ Chromosomes ~~functio~~ condense further during the diakinesis stage, from greek words "meaning through".
- ⇒ This is the first point in meiosis where the four parts of the tetrads are actually visible.
- ⇒ In this stage, the homologous chromosomes separate further, and the chiasmata terminalize. Making chiasmata clearly visible.



2. Metaphase I

- ⇒ Pairs of homologous chromosomes move to the equator of the cell.



3. Anaphase I:

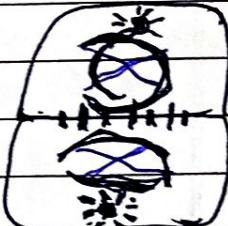
Homozygous chromosomes move to the opposite poles of the cell.



4. Telophase I and Cytokinesis

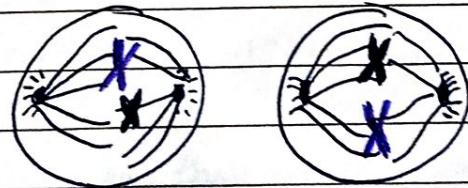
Chromosomes gather at the poles of the cells. The cytoplasm divides.

Sister chromatids arrive at the poles of the cells and begin to decondense. A nuclear envelope forms around each nucleus and the cytoplasm is divided by a cleavage furrow. The result is two haploid cells. Each cell contains one duplicated copy of each homologous chromosome pair.

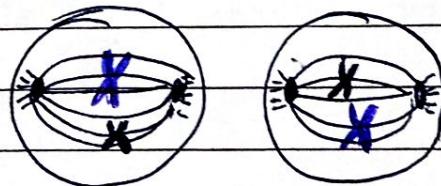


Meiosis II

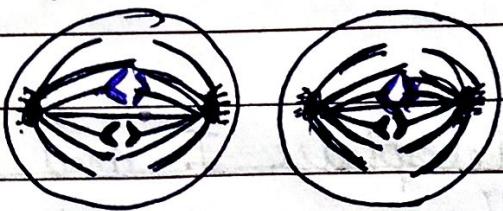
1. Anaphase II: Sister chromatids condense. A new spindle begins to form. The nuclear envelope starts to fragment.



2. Metaphase II: Sister chromatids line up at the metaphase plate.



Anaphase II: Sister chromatids are pulled apart by the shortening of the Kinetochore microtubules. Non-Kinetochore microtubules lengthen the cell.



4. Telophase II and Cytokinesis

Chromosomes arrive at the poles of the cell and decondense. Nuclear envelopes surround the four nuclei. Cleavage furrows divide the two cells into four haploid cells.

