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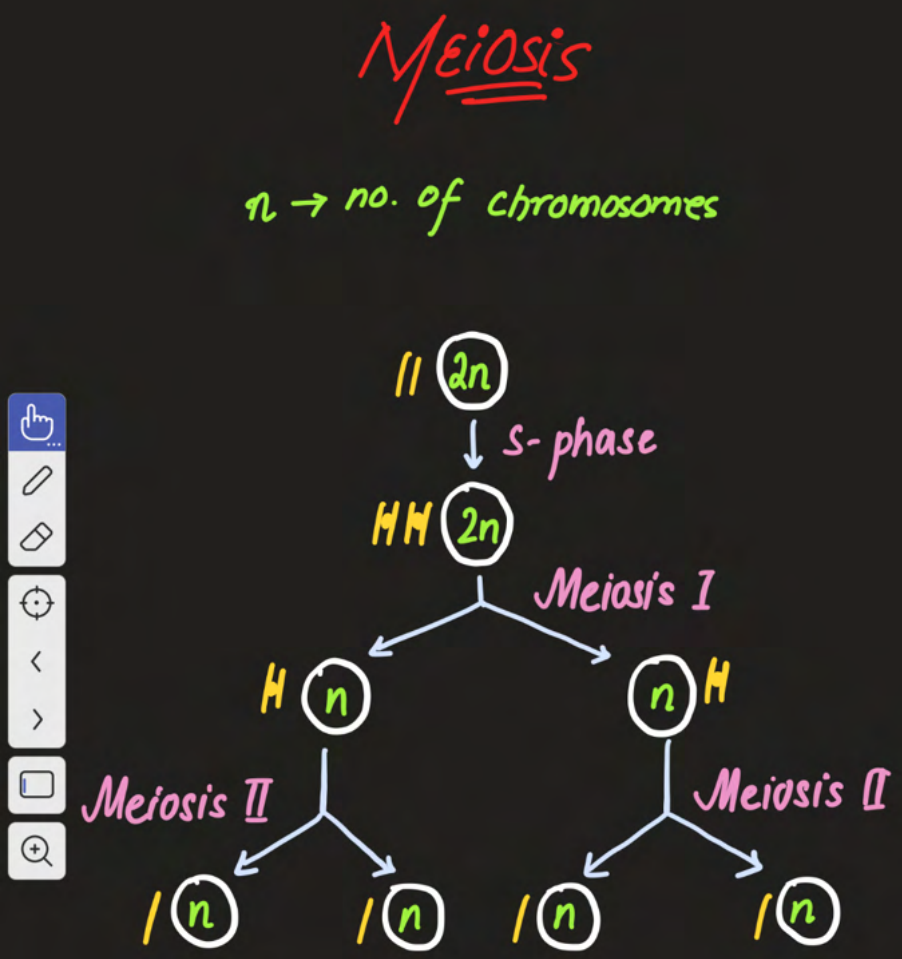
Inherited Change



With
Mohammad Hussham Arshad, MD

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ADVANCED LEVEL BIOLOGY 9700

UNIT 16: Inherited Change

Learning Objectives:

- **Homologous chromosomes**
- **Sister & non-sister chromatids**
- **Overview of meiosis**

Video Lecture 1 Slides

Mohammad Hussham Arshad, MD
Biology Department

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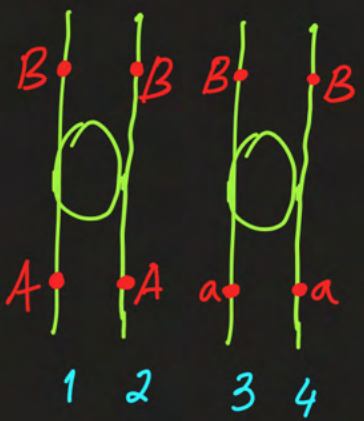
Homologous chromosomes, sister chromatids & non-sister chromatids

A pair of chromosomes is homologous if;

1) same length
2) centromeres at the same position
3) same genes at each loci
4) which may have different alleles



Sister chromatids (genetically identical)

* 1 and 2
* 3 and 4

Non-sister chromatids (genetically non-identical)

* 1 and 3
* 1 and 4
* 2 and 3
* 2 and 4

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HOMOLOGOUS CHROMOSOMES

* Two chromosomes are said to form a homologous pair if:

a) they have the same length

b) their centromeres are at the same position.

c) they have the same genes at the same loci, but may have different alleles.

* A homologous pair of chromosomes may have different alleles of the same gene.

In such a case the organism will be heterozygous for that gene.

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* Sister chromatids are genetically identical chromatids.

* Non-sister chromatids are genetically non-identical.

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Meiosis

* Meiosis

Meiosis I

Meiosis II

* Meiosis I itself is subdivided into:

Prophase 1

Metaphase 1

Anaphase 1

Telophase I

* Meiosis II itself is subdivided into:

Prophase II

Metaphase II

Anaphase II

Telophase II

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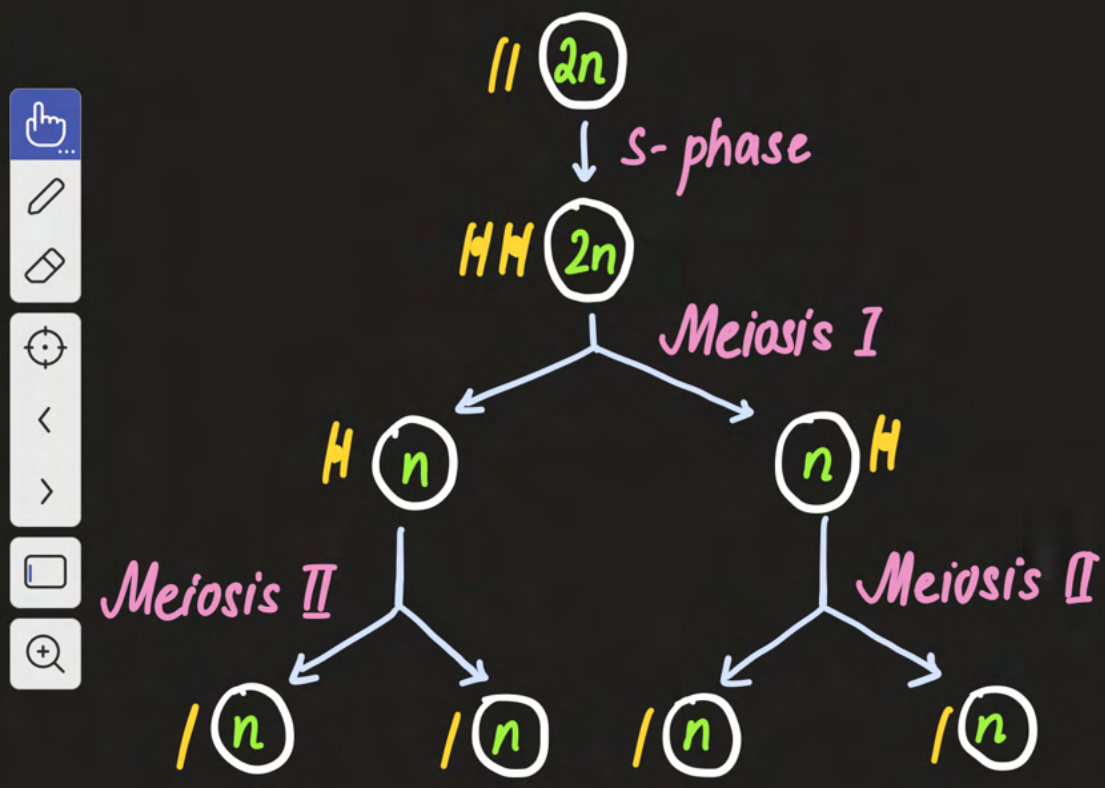
Meiosis

n → no. of chromosomes



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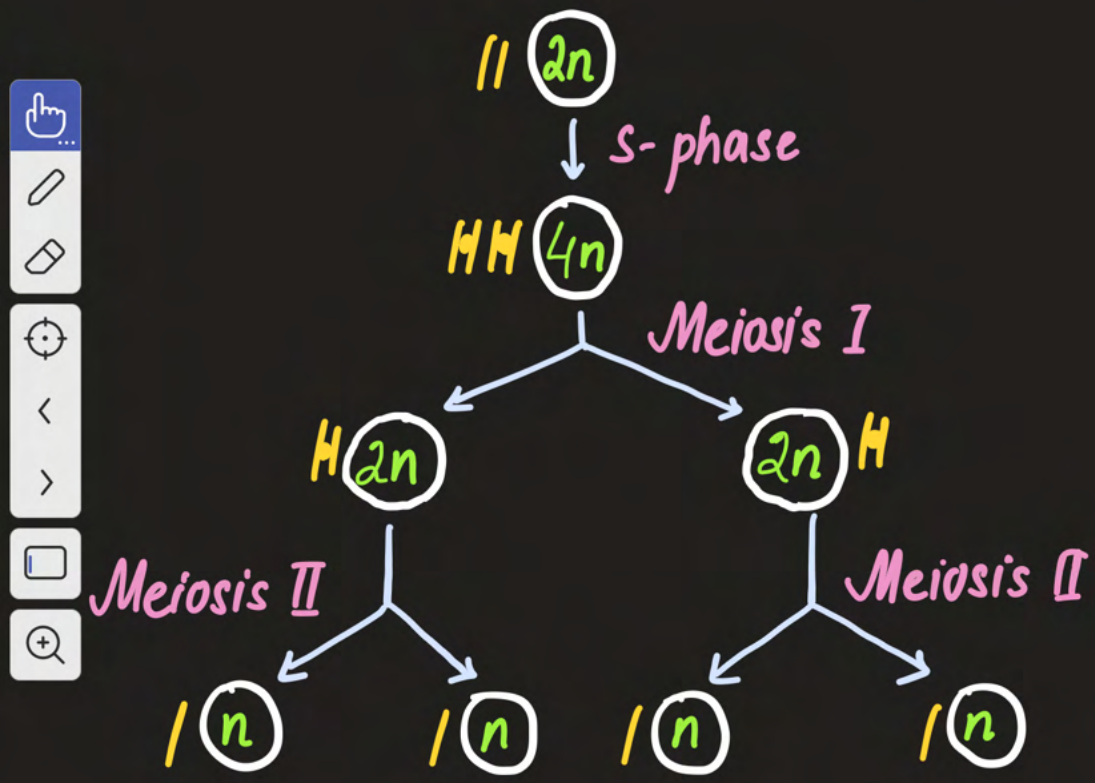
Meiosis

n → no. of chromatids / DNA copies



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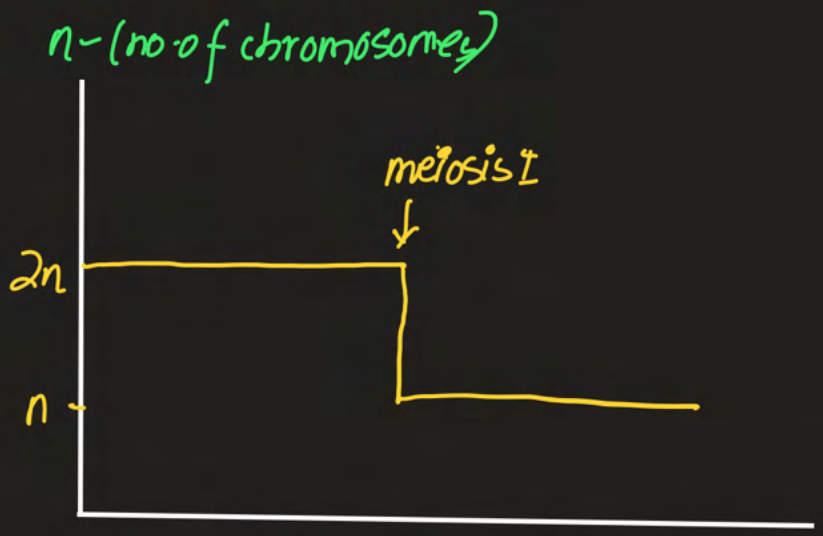
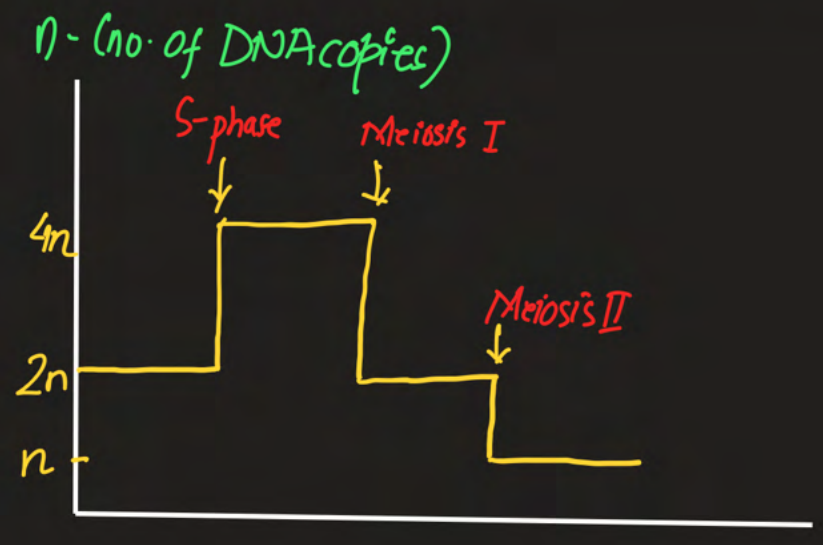
Meiosis





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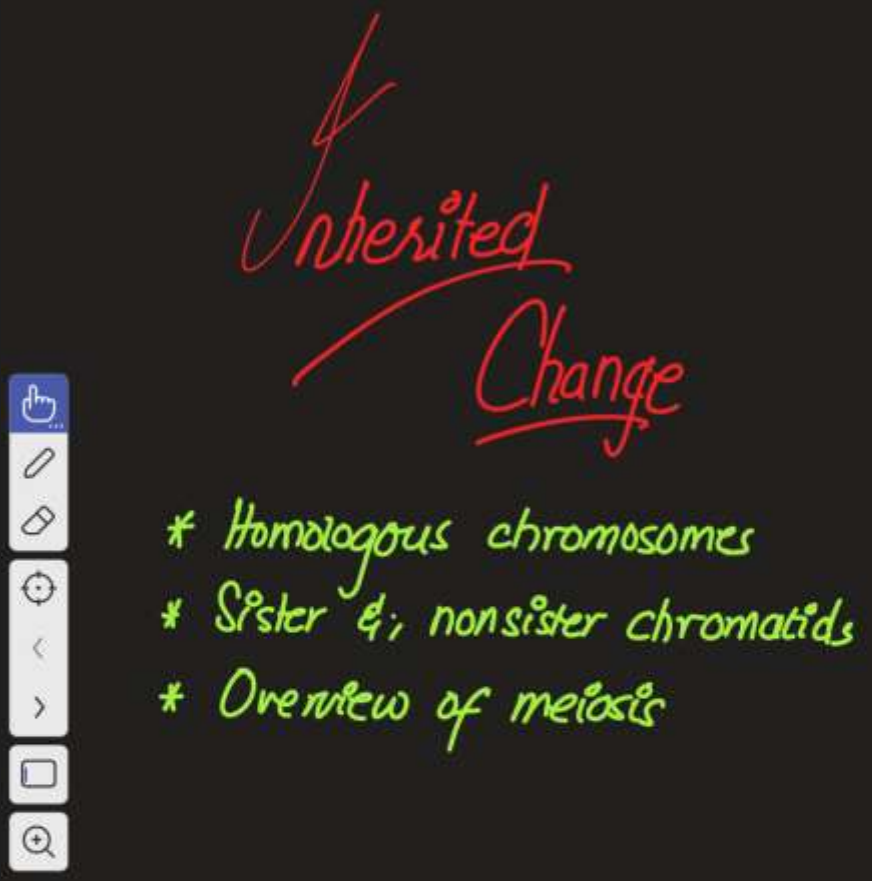
Inherited Change



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ADVANCED LEVEL BIOLOGY 9700

UNIT 16: Inherited Change

Learning Objectives:

- Meiosis 1 & 2
- Meiosis & genetic variation
- Fertilisation & genetic variation

Video Lecture 2 Slides

Mohammad Hussham Arshad, MD
Biology Department

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Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis

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Meiosis

* Meiosis

Meiosis I

Meiosis II

* Meiosis I itself is subdivided into:

Prophase 1

Metaphase 1

Anaphase 1

Telophase I

* Meiosis II itself is subdivided into:

Prophase II

Metaphase II

Anaphase II

Telophase II

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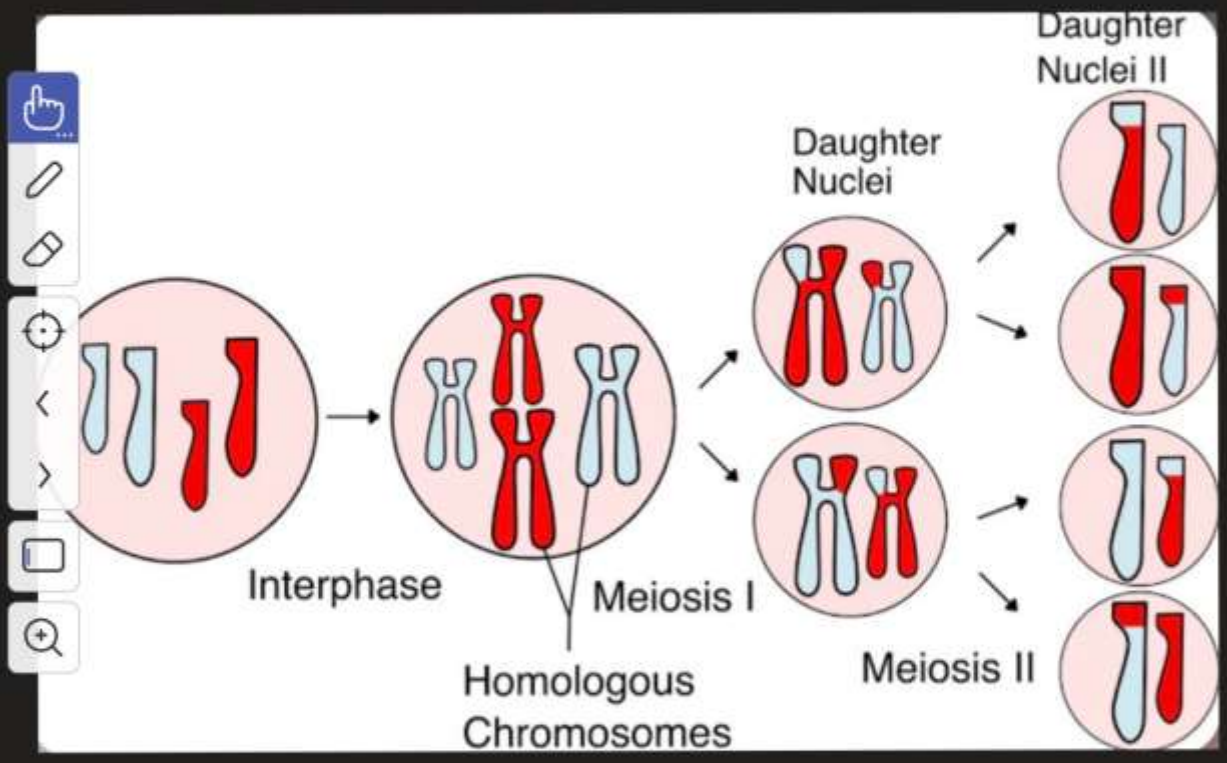
Daughter
Nuclei II
Daughter
Nuclei
Interphase
Meiosis I
Homologous
Chromosomes
Meiosis II

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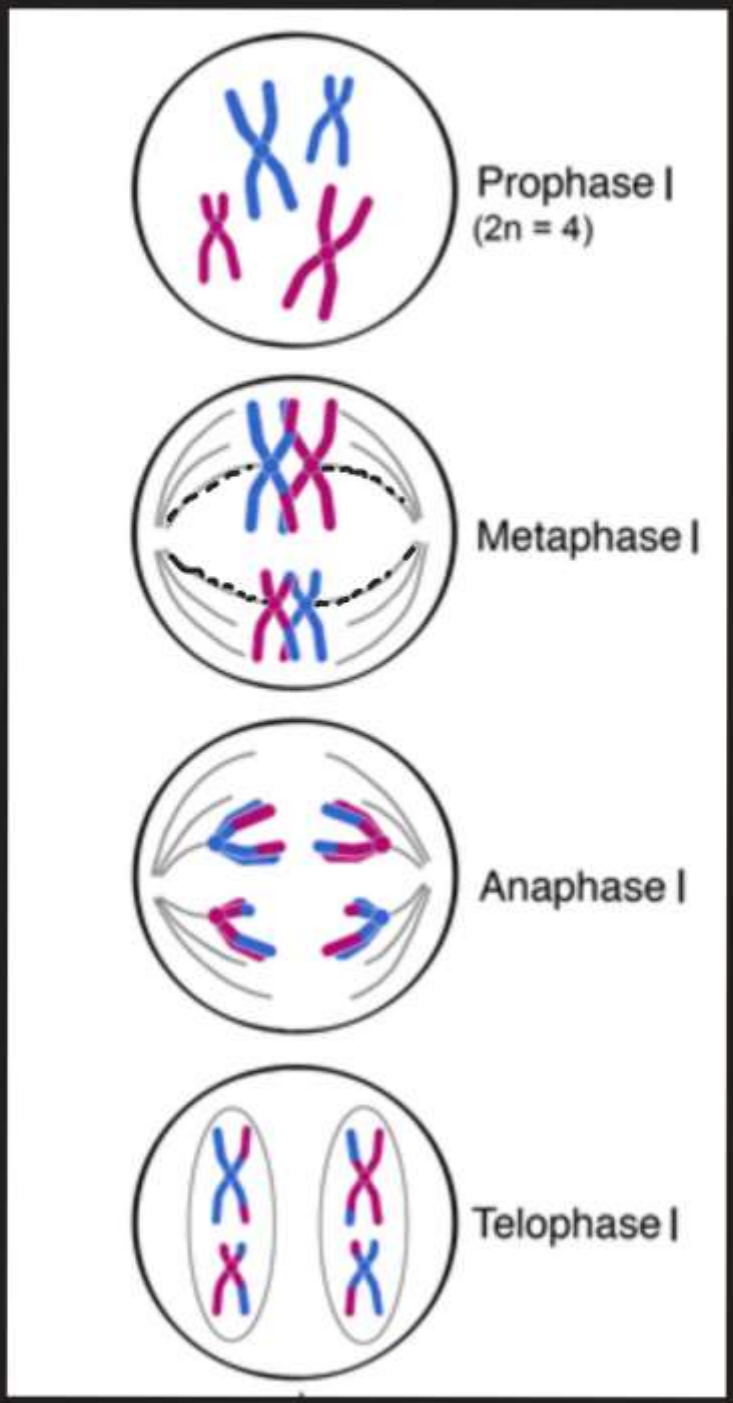
Meiosis I



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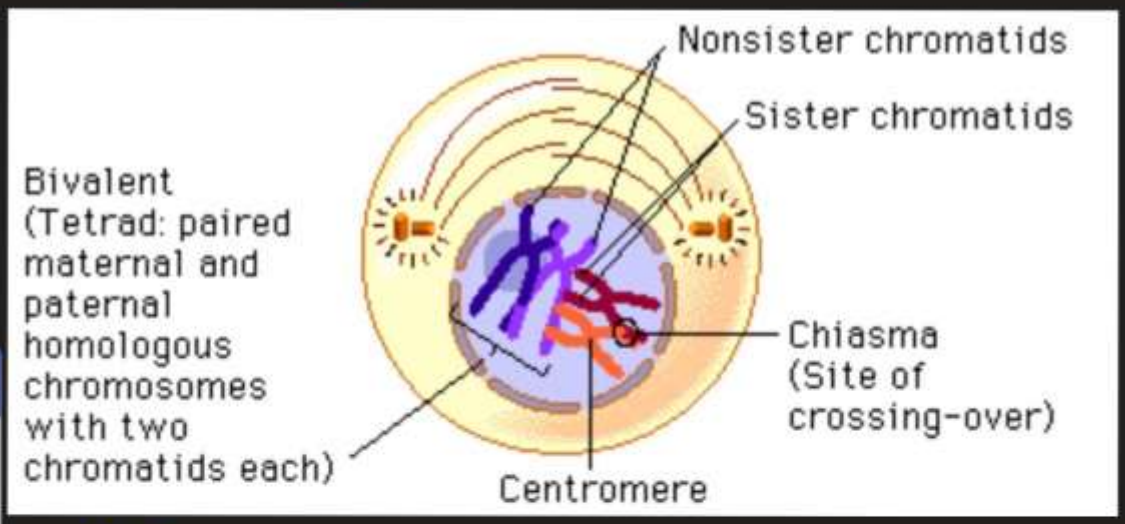
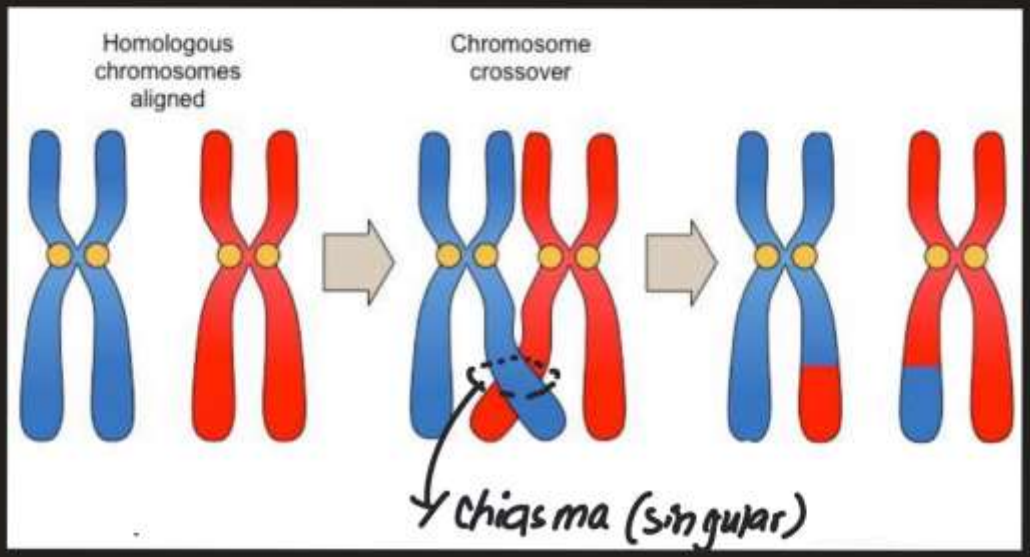
Meiosis I → Prophase I



1) Homologous chromosomes pair up → synapsis → to form bivalents (tetrads)
2) Chiasmata formation



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Meiosis I

Prophase I:

* The chromosomes shorten and thicken
* Homologous chromosomes pair up. This process is called **synapsis**. Each pair is termed as a **bivalent**.
* The centrosomes migrate towards the opposite poles of the spindle.
* The nuclear envelope disintegrates.
* The nucleolus disappears.
* Spindle formation occurs
* Chiasmata forms
* Crossing over may occur

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Chiasma formation:

* Chiasma form, each chiasma is a region of overlap between non-sister chromatids.

Chiasmata formation is important for the following reasons:

a) It holds the Two chromosomes within a bivalent together. (mechanical role).

b) Crossing over may occur due to overlap b/w non-sister chromatids (genetic role)

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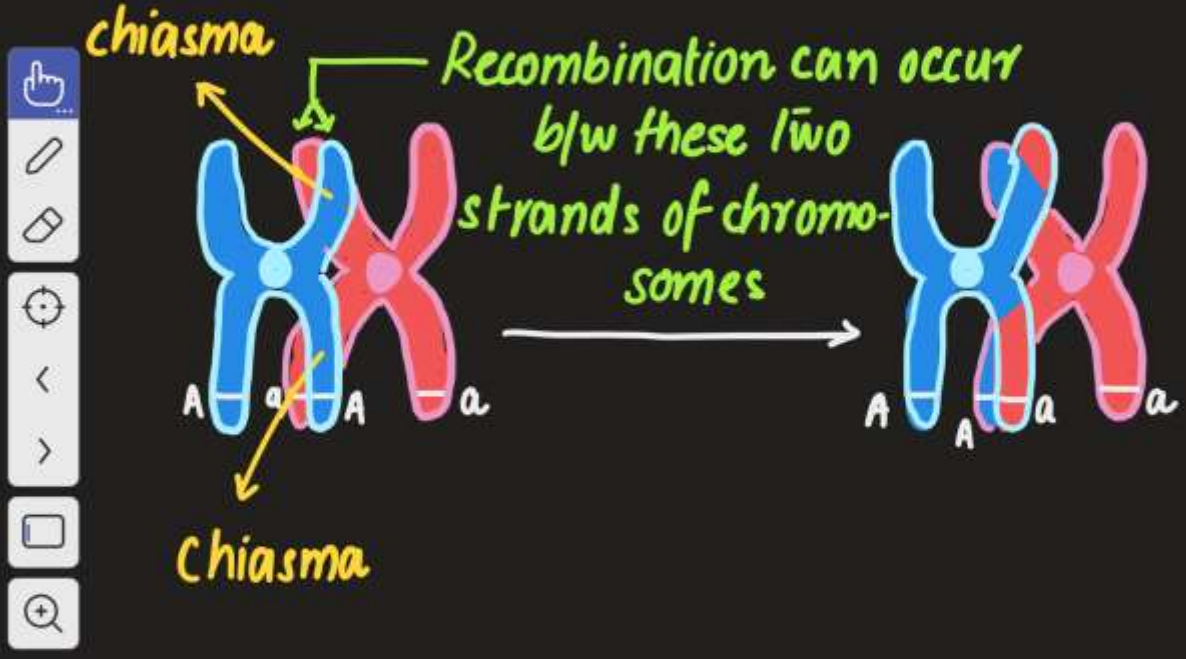
chiasma
Recombination can occur b/w these two strands of chromo-somes
A
a
A
a
A
A
a
a
Chiasma

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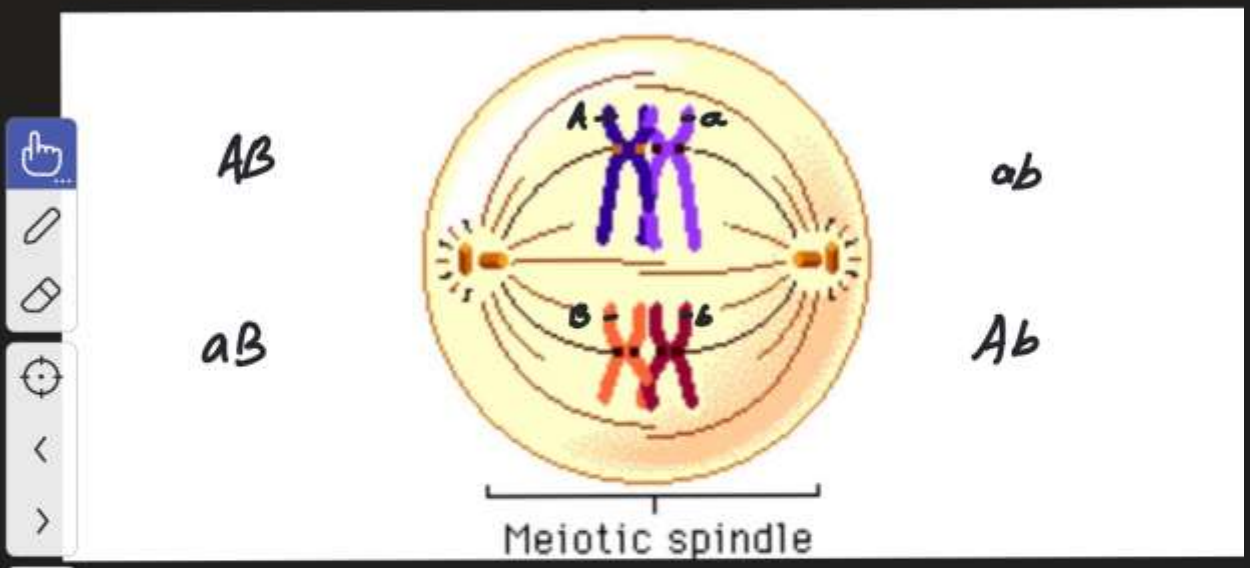
Meiosis I → metaphase I



n → no. of bivalents

2^n → different types of gametes

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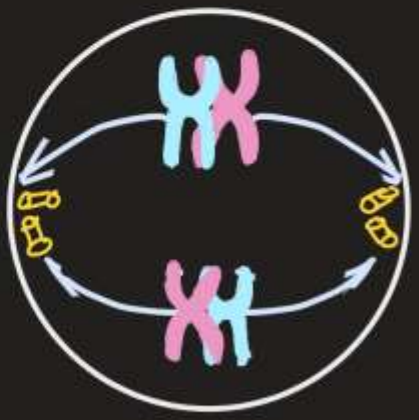
Metaphase I :

* Homologous pairs of chromosomes align themselves along the equator of the spindle.
* Chromosomes are attached to the spindle fibres via kinetochores.
* Independent assortment occurs.



Metaphase I

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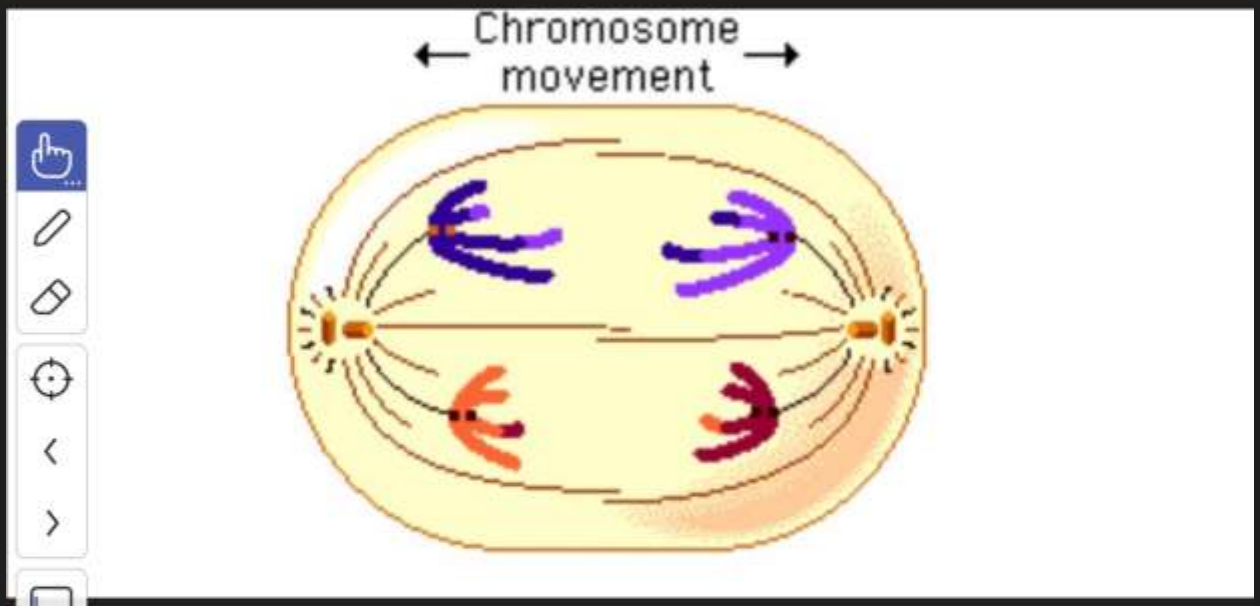
Meiosis I → Anaphase I



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Anaphase 1

* Individual chromosomes move towards the opposite poles of the spindle with centromeres leading.
* Centromeres do not divide.
* The movement of the chromosomes occurs due to the contraction of the microtubules.
* Chromosomes detach from the spindle fibres at the end of anaphase.

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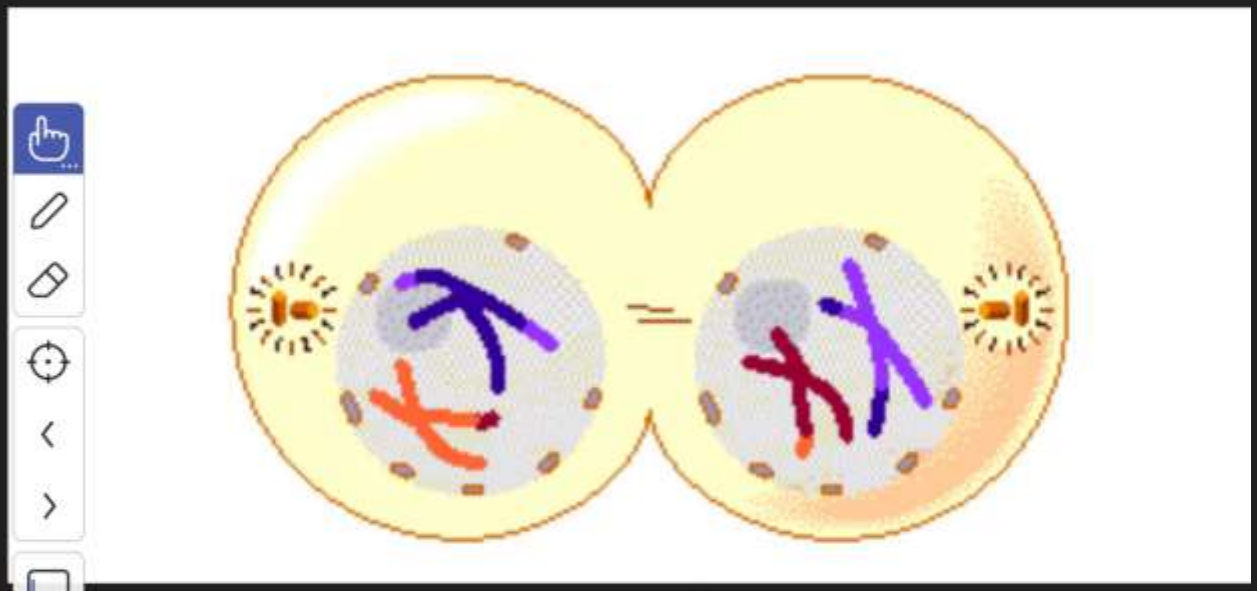
Meiosis I → Telophase I



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Telophase I

* The nuclear envelope reforms
* The nucleolus reappears.
* The chromosome number reduces from 2n To n.
* Animal cells enter cytokinesis before Meiosis II.
* Plant cells enter Meiosis II without cytokinesis.

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Meiosis II

- Prophase II
- Metaphase II
- Anaphase II
- Telophase II

} Chromosomal behaviour is VERY similar to Mitosis

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Meiosis II

1) Prophase II

→ Chromosomes shorten and thicken (condensation)

→ Centrosomes migrate to the opposite poles of the nucleus.

→ Nuclear envelope disintegrates.

→ Nucleolus disappears

→ Spindle formation occurs.

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Metaphase II

→ Chromosomes attach to the spindle fibres via kinetochores.

→ Chromosomes are aligned along the equator of the spindle.

Metaphase I

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Anaphase II :

→ Centromere divides

→ chromatids move towards the opposite poles of the spindle with centromeres leading.

→ The movement of chromatids is due to contraction of microtubules.

→ Chromatids detach from spindle fibres at the end of anaphase.

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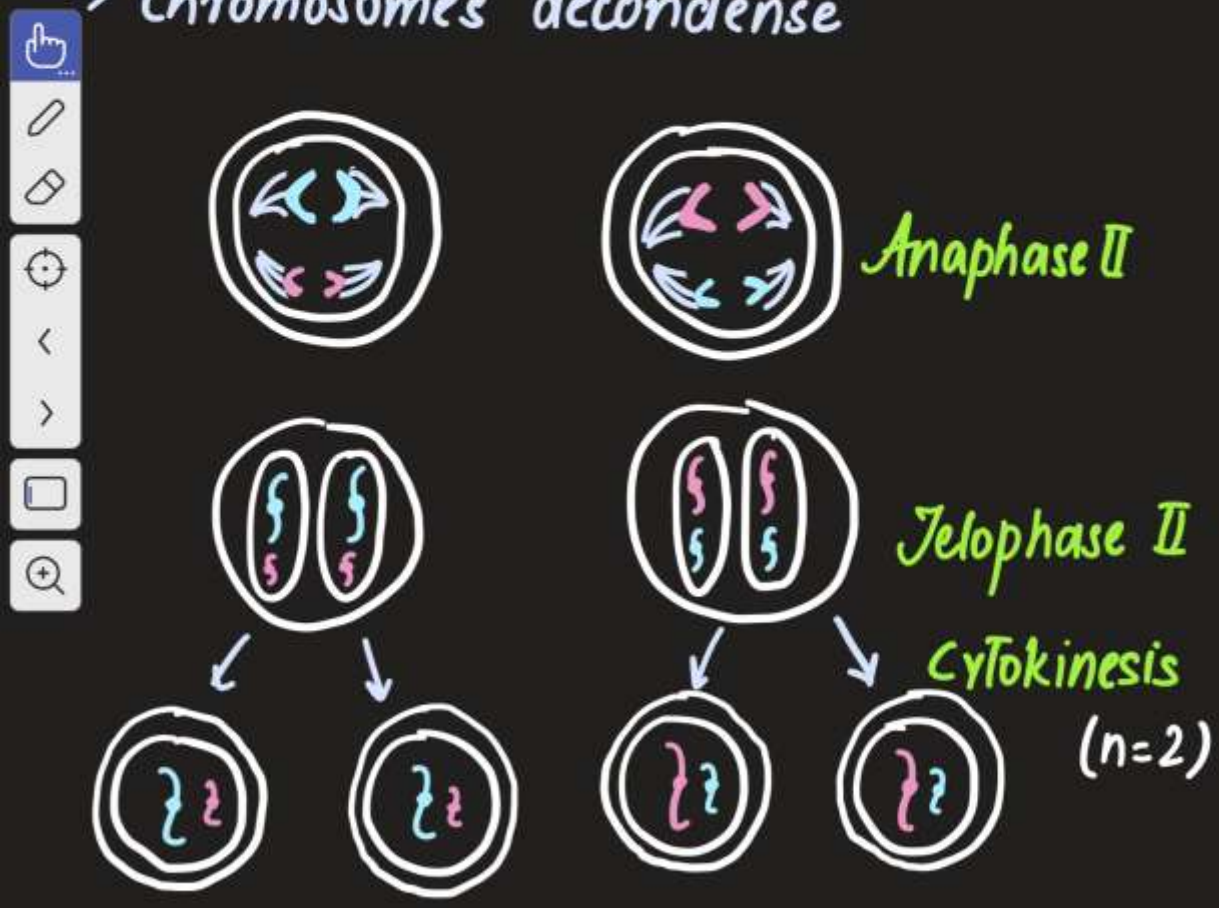
Telophase II

→ Nuclear envelope reforms.

→ Nucleolus reappears

→ Chromosomes decondense



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Meiosis and Genetic variation

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Meiosis and genetic variation

*Meiosis can lead to genetic variation in the following ways:

- Crossing over
- Independent Assortment
- Mutation

LINKED AND UNLINKED ALLELES

*Alleles that are present on the same chromosome are said to be LINKED (linkage)

*Alleles that are present on different chromosomes are said to be UN-LINKED (non-linkage)

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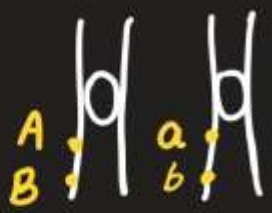
* Consider the following pair of homologous chromosomes with the genes A/a & B/b:



NOTE: ① allele A is linked to allele B

② allele a is linked to allele b.

③ alleles a and b are unlinked to alleles A and B.

④ Linked alleles are inherited together

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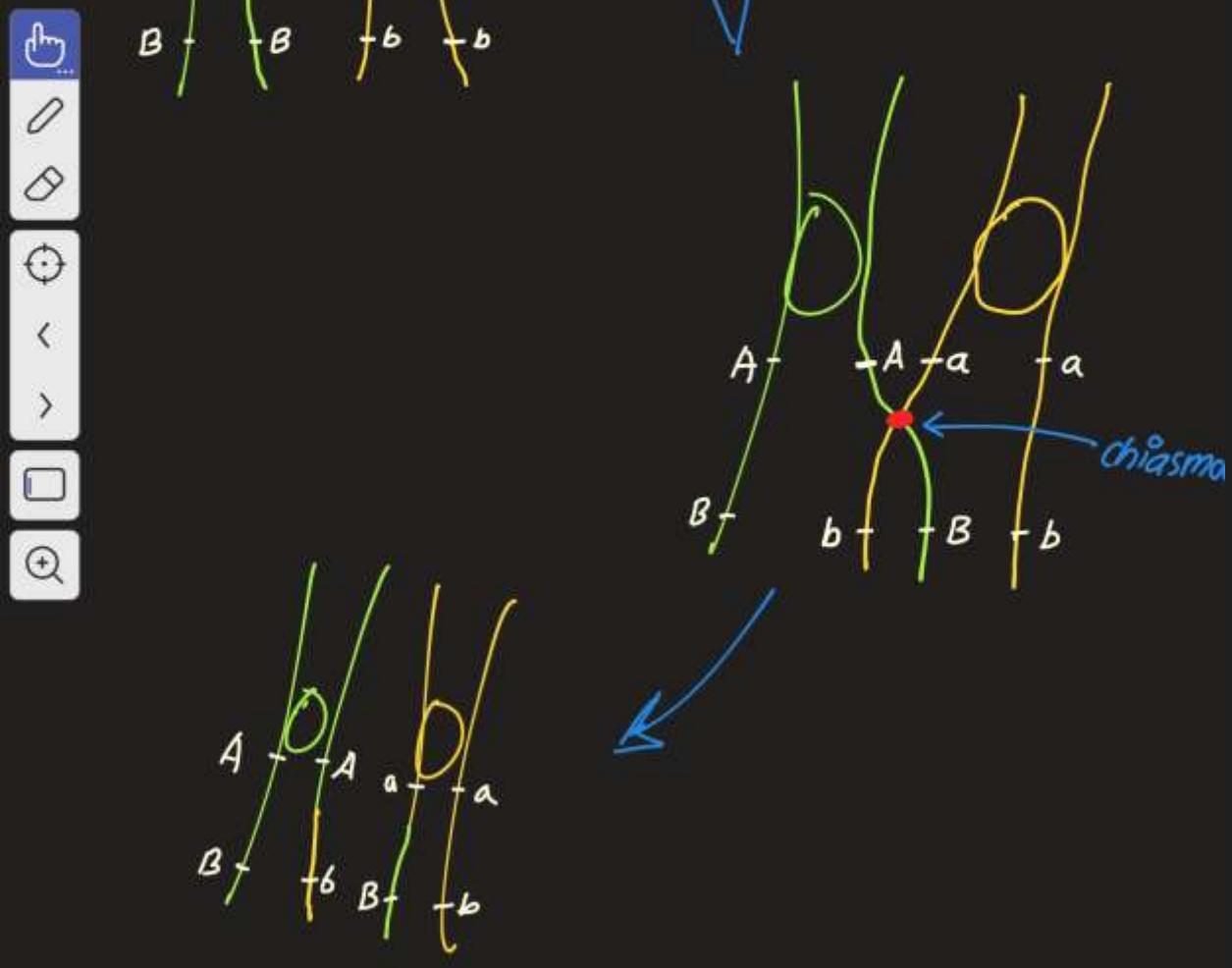
Crossing Over



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CROSSING OVER

*It is defined as The exchange of genetic material b/w non-sister chromatids during prophase I of meiosis I.

* Crossing over occurs due to The formation of chiasmata b/w non-sister chromatids.

*Crossing over leads to genetic variation due to breakage of linkage groups leading to formation of new combination of alleles.

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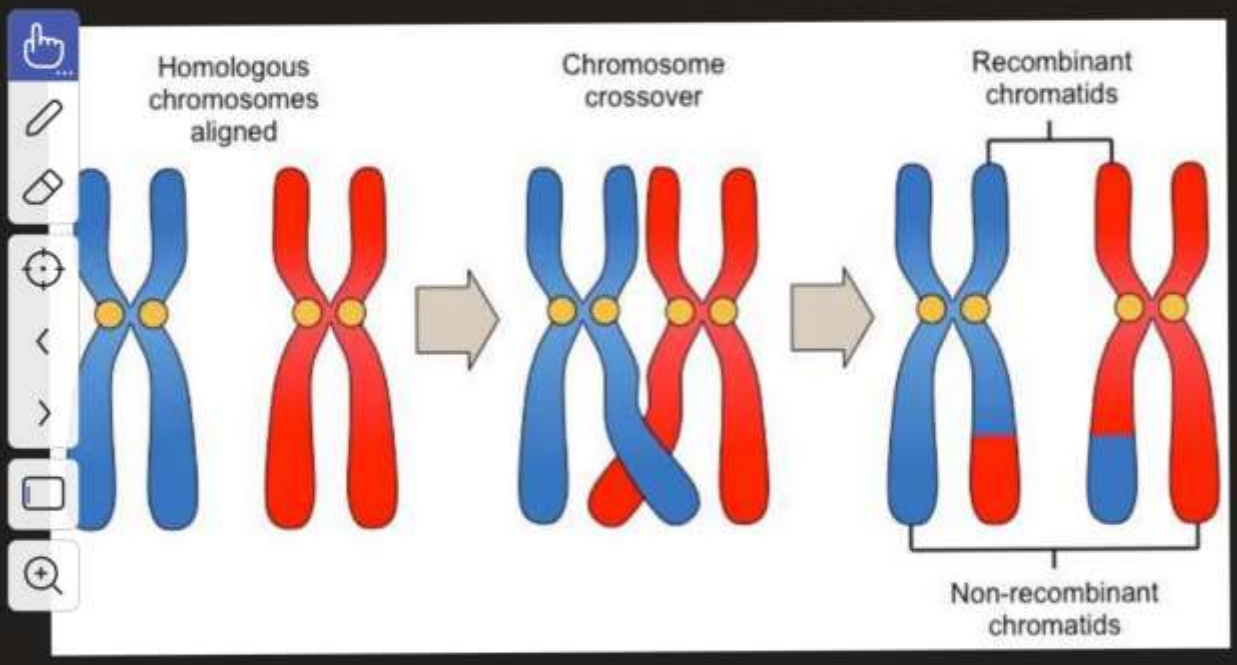
Homologous chromosomes aligned
Chromosome crossover
Recombinant chromatids
Non-recombinant chromatids

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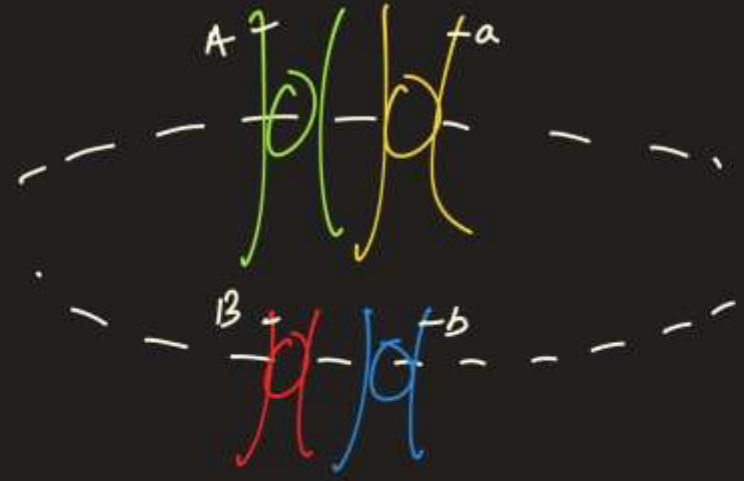
Independent Assortment

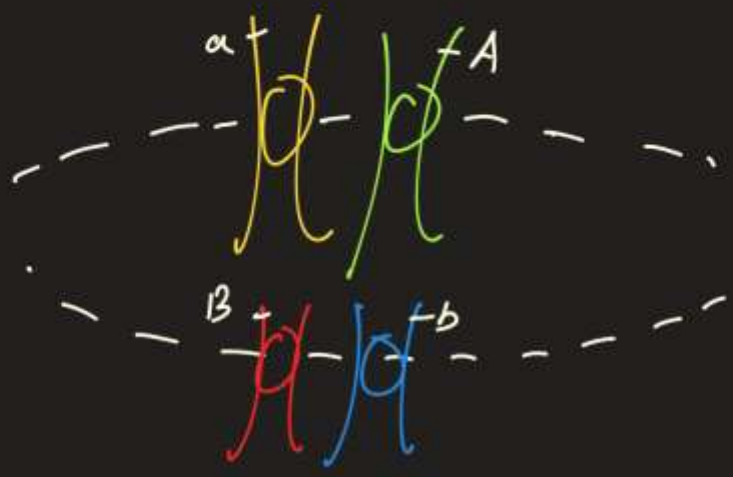
AB

ab



aB

Ab



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Independent Assortment

* Independent assortment is defined as the random orientation of the homologous chromosomes along the equator of the spindle during Metaphase of Meiosis.
Each bivalent aligns itself independently of others.
* Independent assortment may lead to genetic variation by forming 2^n possible number of different gametes where n is the number of bivalents with a particular nucleus.

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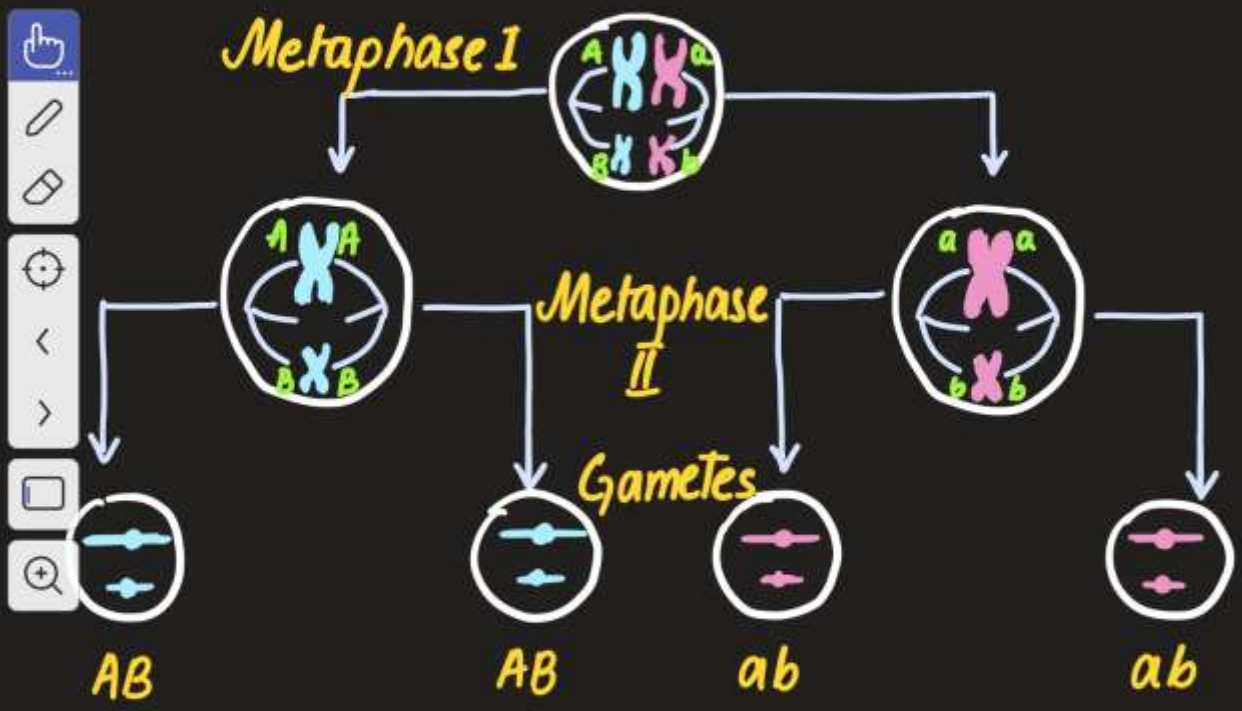
* Since human germ cells form 23 bivalents, the total possible number of different gametes that can be produced are = 2^{23}



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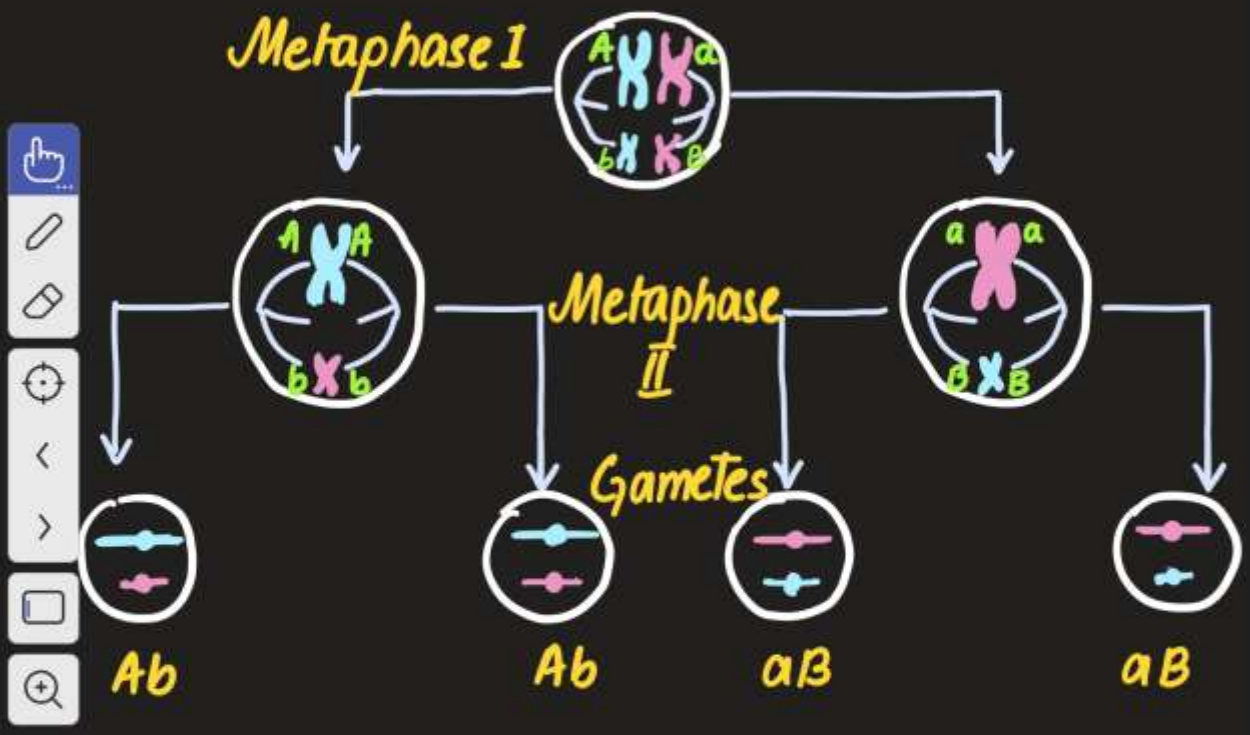

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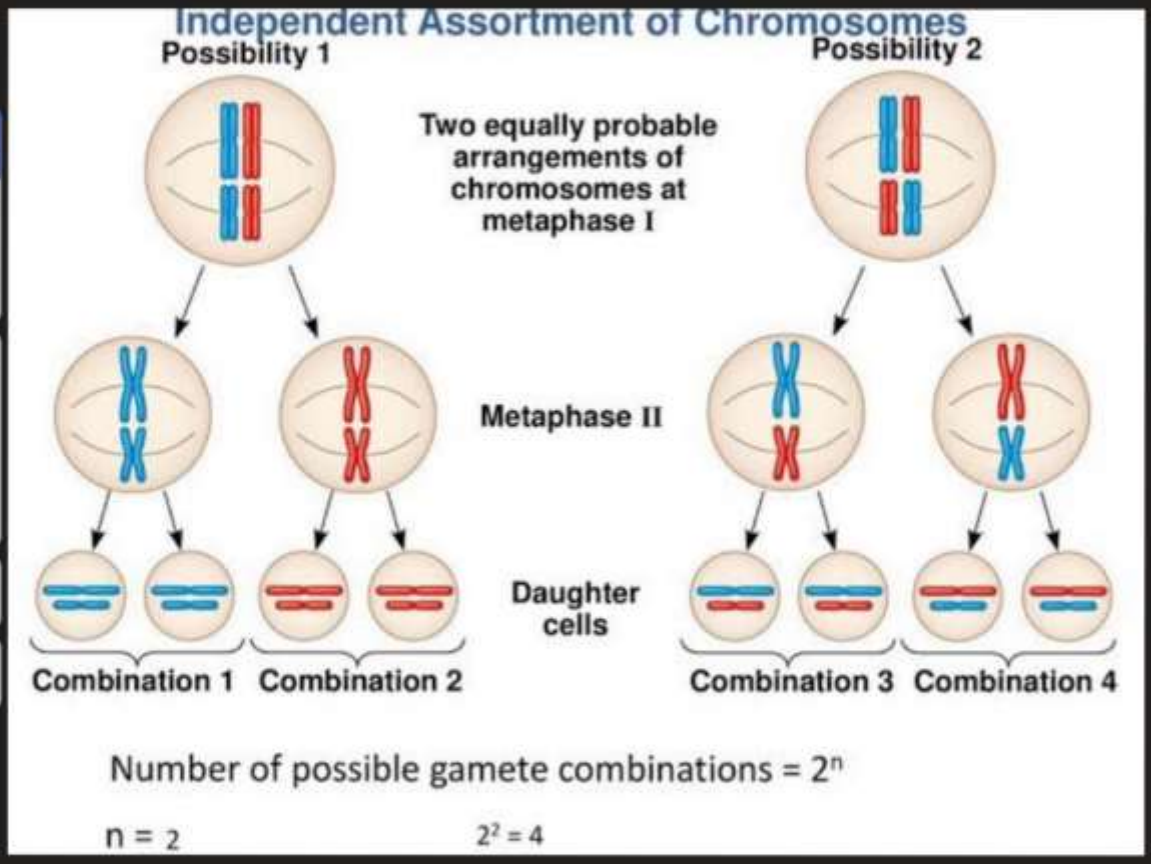
Independent Assortment of Chromosomes
Possibility 1
Possibility 2
Two equally probable arrangements of chromosomes at metaphase I
Metaphase II
Daughter cells
Combination 1
Combination 2
Combination 3
Combination 4
Number of possible gamete combinations = 2^n
n = 2
$2^2 = 4$

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Mutation

*It is described as the alter ation in the sequence of nucleotides or the chromosome number or structure.

* It leads to variation by forming new alleles.

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Fertilisation and genetic variation

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Fertilisation as a source of genetic variation

* It is defined as the fusion of 2 haploid gametes to produce a diploid cell for example a human sperm cell may fuse with ovum to produce a diploid zygote.

* It may lead to genetic variation because the fusion of gametes is random.

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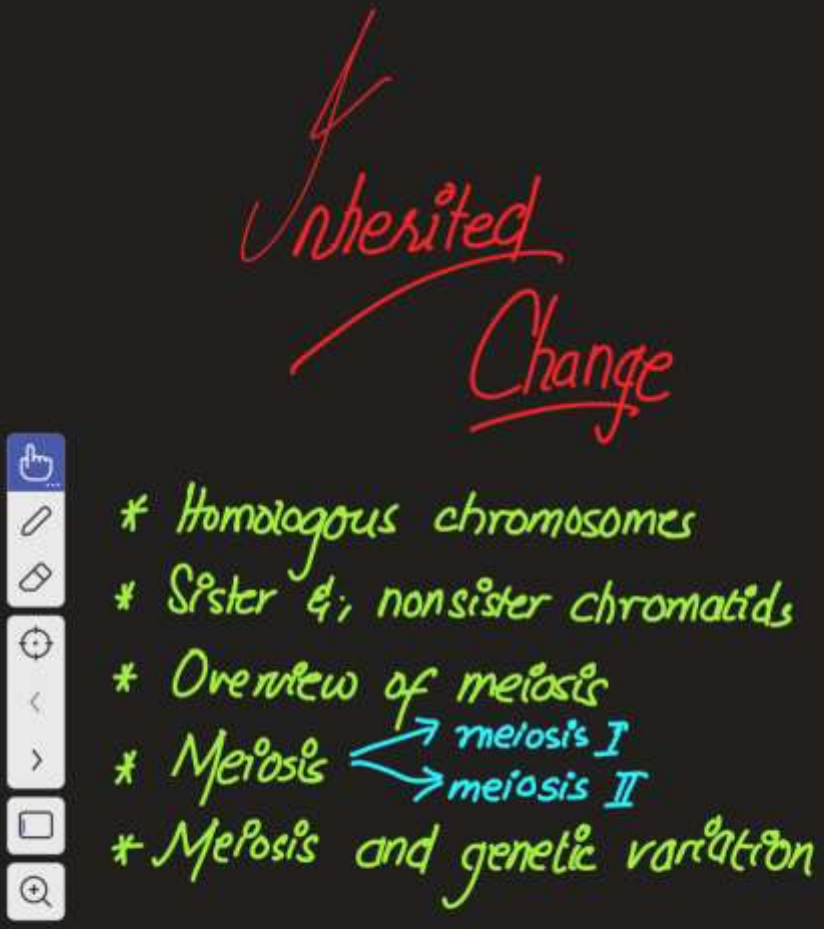
Inherited Change



With
Mohammad Hussham Arshad, MD

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ADVANCED LEVEL BIOLOGY 9700

UNIT 16: Inherited Change

Learning Objectives:

- **Differences between mitosis & meiosis**
- **Overview of gametogenesis**
- **Gametogenesis in mammals: spermatogenesis & oogenesis**

Video Lecture 3 Slides

Mohammad Hussham Arshad, MD
Biology Department

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Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis
* Meiosis → meiosis I, meiosis II
* Meiosis and genetic variation

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Q1: Differentiate between mitosis and meiosis?

Mitosis	Meiosis
• Conserves chromosome number.	• halves the chromosome number
• ensures genetic uniformity	• leads to genetic variation.
• Homologous chromosomes do not pair up during prophase.	• Homologous chromosomes pair up to form bivalents in prophase I.
• Chiasmata formation does not occur in prophase	• Chiasmata formed in prophase I which may lead to crossing over.

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Mitosis	Meiosis
• Individual chromosomes line across the equator of the spindle with no independent assortment in metaphase.	• Bivalents line along the equator of the spindle. Independent assortment occurs in metaphase I.
• Centromeres divide in anaphase of mitosis.	• No centromere division in anaphase I of meiosis

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Mitosis	Meiosis
• Sister chromatids move towards the opposite poles of the spindle in anaphase.	• chromosomes move towards the opposite poles of the spindle in anaphase I.
• may form haploid or diploid cells.	• meiosis leads to formation of haploid cells from diploid cells.

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Q_2: Outline how meiosis leads to genetic variation?

Ans: Meiosis is a form of reduction division which leads to genetic variation through crossing over and independent assortment. Crossing over refers to the exchange of genetic material between non-sister chromatids of a bivalent. It occurs during prophase I of meiosis. It leads to genetic variation by forming new combinations

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of alleles due to breakage of linkage groups. Independent assortment refers to the random alignment of bivalents across the equator of the spindle during metaphase. Each bivalent lines up independently of others. It leads to genetic variation by forming 2^n possible numbers of different gametes where n is the no. of bivalents.

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Q3.

4 Fig. 4.1 shows the results of an animal cell that has undergone meiosis I.

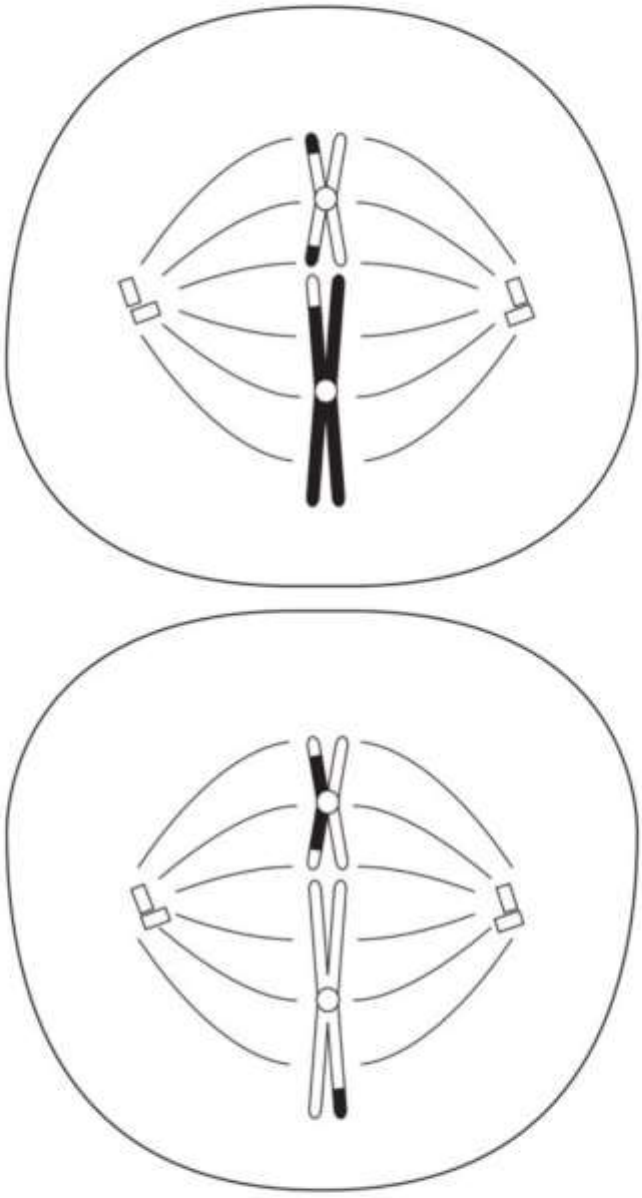

Fig. 4.1

(a) State which stage of meiosis II is shown.

metaphase II ..... [1]

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(b) Describe the next stage of meiosis II.

* centromere divides
* chromatids move towards the opposite poles of the spindle
* with centromeres leading
* movement due to contraction of the micro-tubules [3]

(c) Describe what has happened before the start of meiosis to

(i) the nuclear membrane;

* disintegrates / breaks down [1]

(ii) the centrioles.

* replicate and
* move towards the opposite poles of the spindle [2]

(d) Name and explain two ways in which meiosis can lead to variation.

1. Crossing over due to exchange of genetic material bw non-sister chromatids. This leads to formation of new combination of alleles.
2. Independent assortment which refers to random alignment of the bivalents along the equator. It leads to formation of 2^n possible no. of gametes. [4]

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Q4.

7 Meiosis is a type of nuclear division, which produces gametes for sexual reproduction.

(a) Fig. 7.1 shows diagrams of the stages of meiosis, **A** to **J**, but they are not in the correct order.

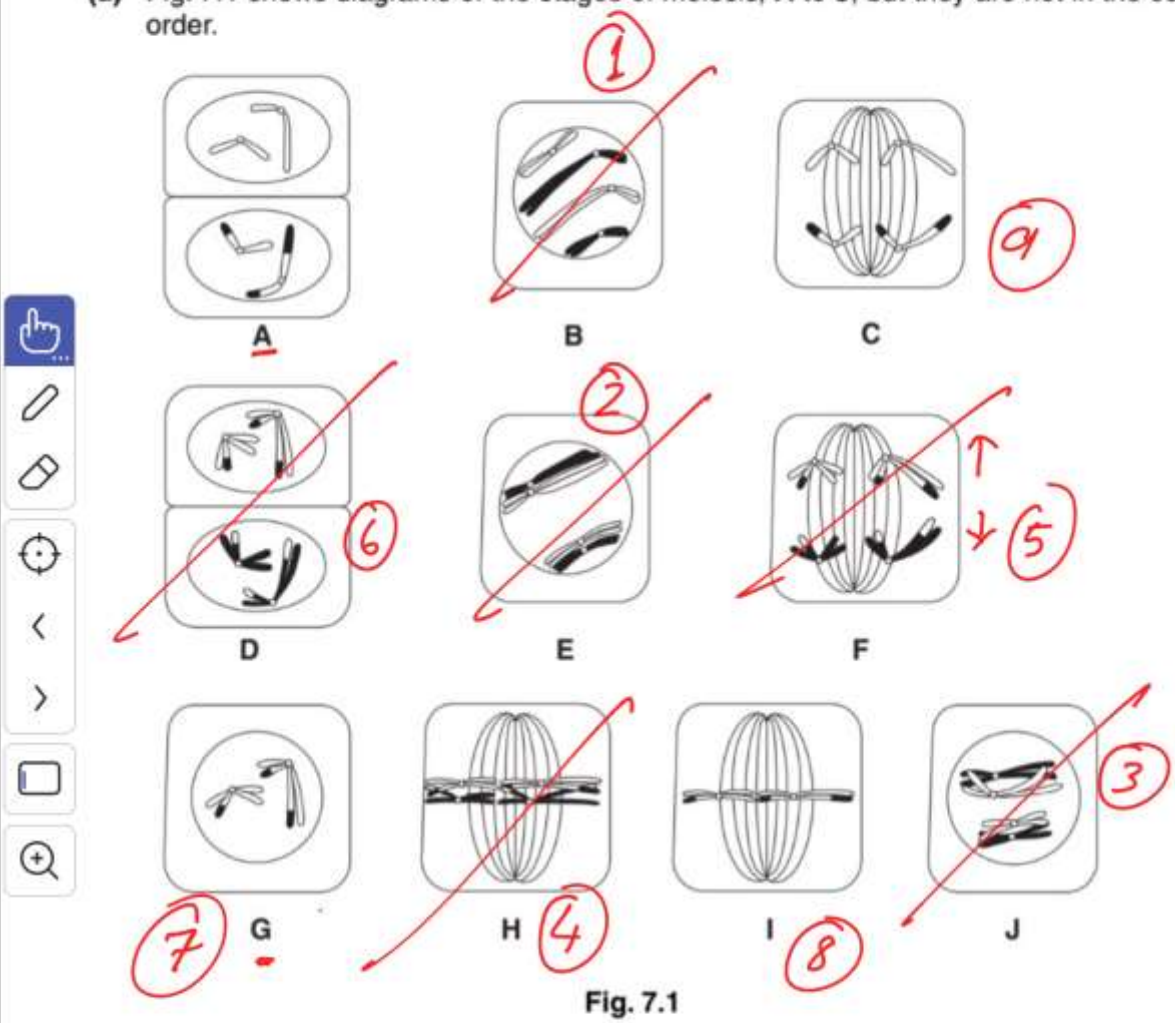

Fig. 7.1

Meiosis I B → E → J → H → F → D

Meiosis II G → I → C → A

A C G I

Complete the table below by writing the stages of meiosis in the correct order.

Some of the stages have already been written in the table.

nuclear division	letter of stage
meiosis I	B
	E
	J
	H
	F
	D
meiosis II	G
	I
	C
	A

[4]

(b) Explain how meiosis can result in genetic variation amongst offspring.

* Crossing over ✓

* Indep. assortment ✓

* mutation ✓

[5]

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Q5.

2 Fig. 2.1 is a diagram of pair of homologous chromosomes during meiosis.

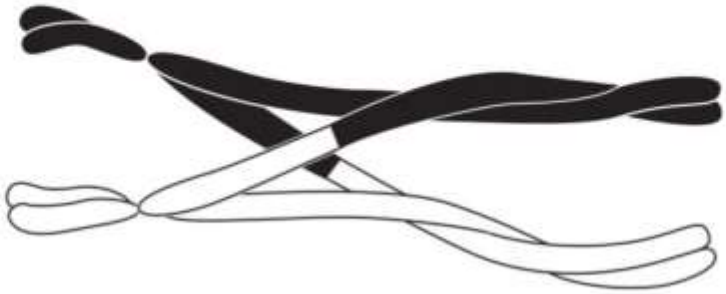

Fig. 2.1

(a) State what stage of meiosis is shown.

late prophase I ..... [1]

(b) Describe what has occurred between the two homologous chromosomes.

Crossing over has occured which involves exchange of genetic material b/w non-sister chromatids.

..... [3]

(c) Explain how this can lead to variation.

Crossing over leads to formation of new combination of alleles due to breakage of linkage groups ..... [2]

(d) Describe **two** other sources of variation that are possible as a result of meiosis.

* Independent assortment

* Mutation

..... [4]

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GAMETOGENESIS

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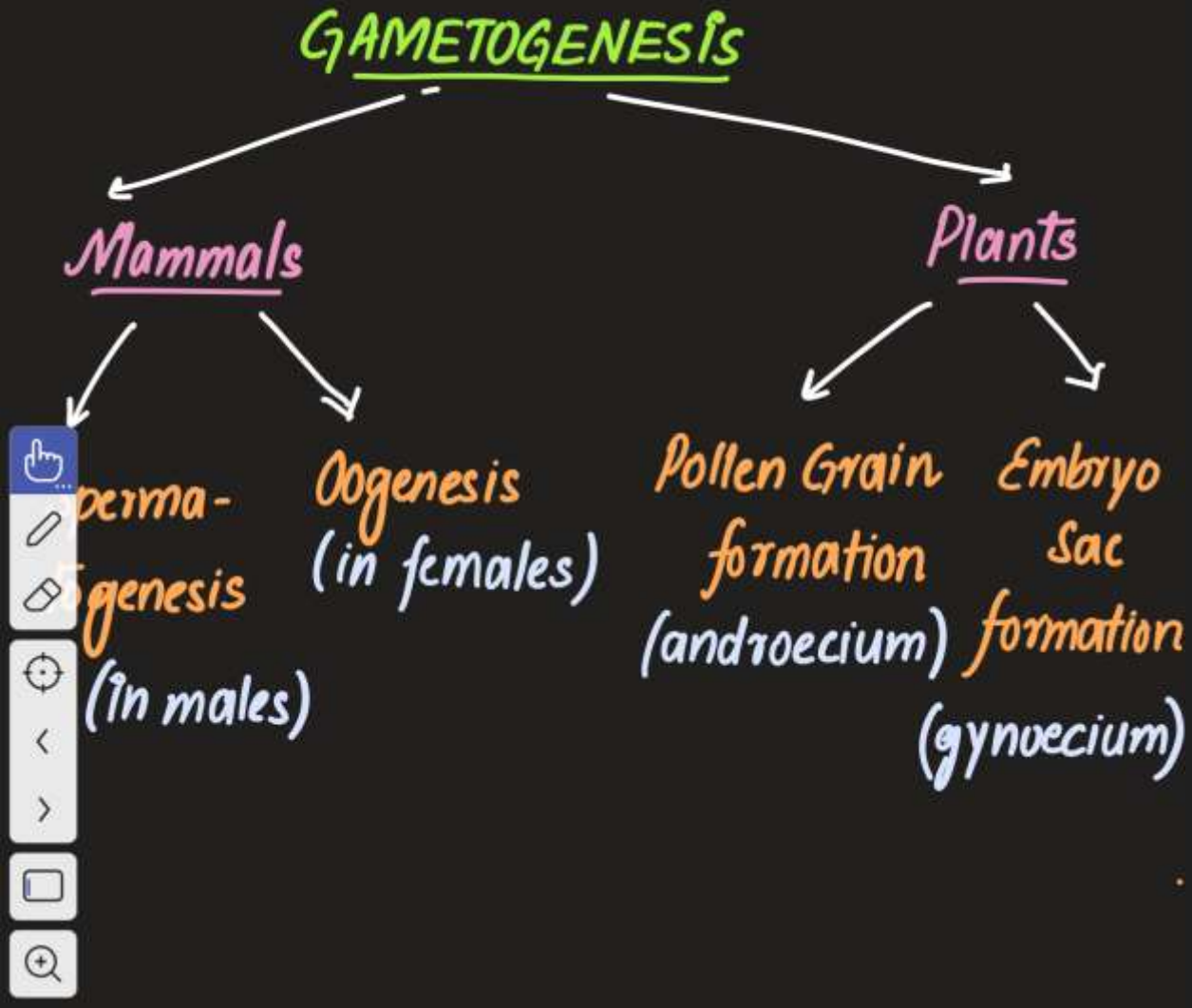
GAMETOGENESIS
Mammals
Plants
perma-
genesis
(in males)
Oogenesis
(in females)
Pollen Grain
formation
(androecium)
Embryo
Sac
formation
(gynoecium)

HEUSER

Meiosis in mammals

* Meiosis leads to formation of gametes in males and females.
* The process that leads to formation of gametes is known as gametogenesis.
* Gametogenesis exists in two forms in mammals:

1. Spermatogenesis
2. Oogenesis

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Spermatogenesis

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Spermatogenesis:

* Refers to the production of haploid spermatozoa from diploid spermatogonia.
* It occurs in males.
  - The site of spermatogenesis are the testes
  - The germinal epithelium of the seminiferous tubules contains numerous diploid spermatogonia which serve as germ cells to form haploid spermatozoa.

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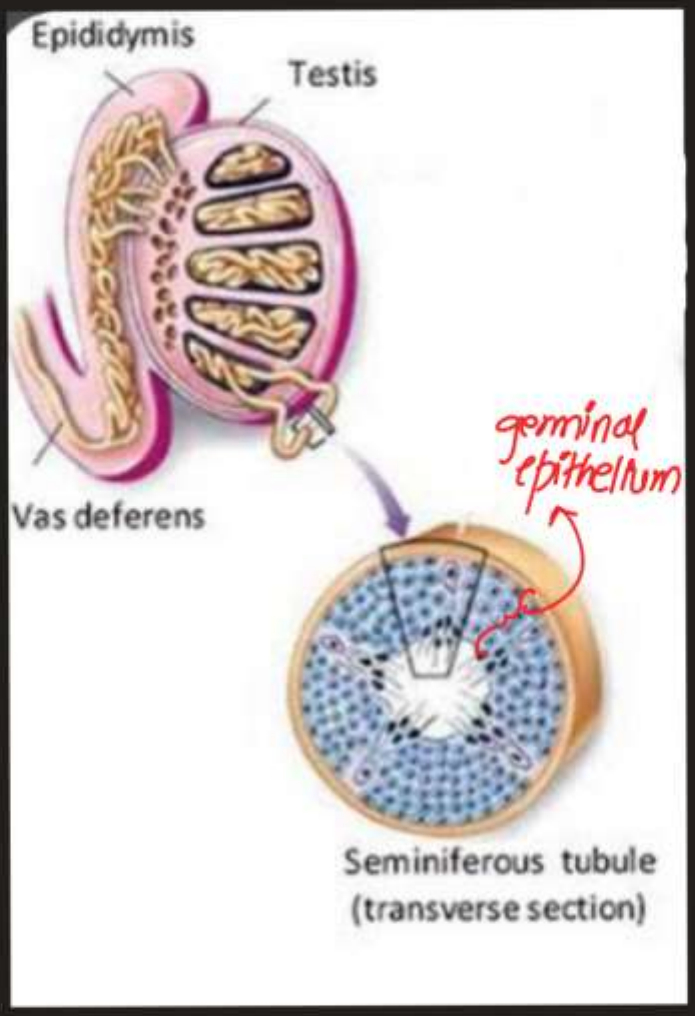
Testis, seminiferous tubules & the germinal epithelium



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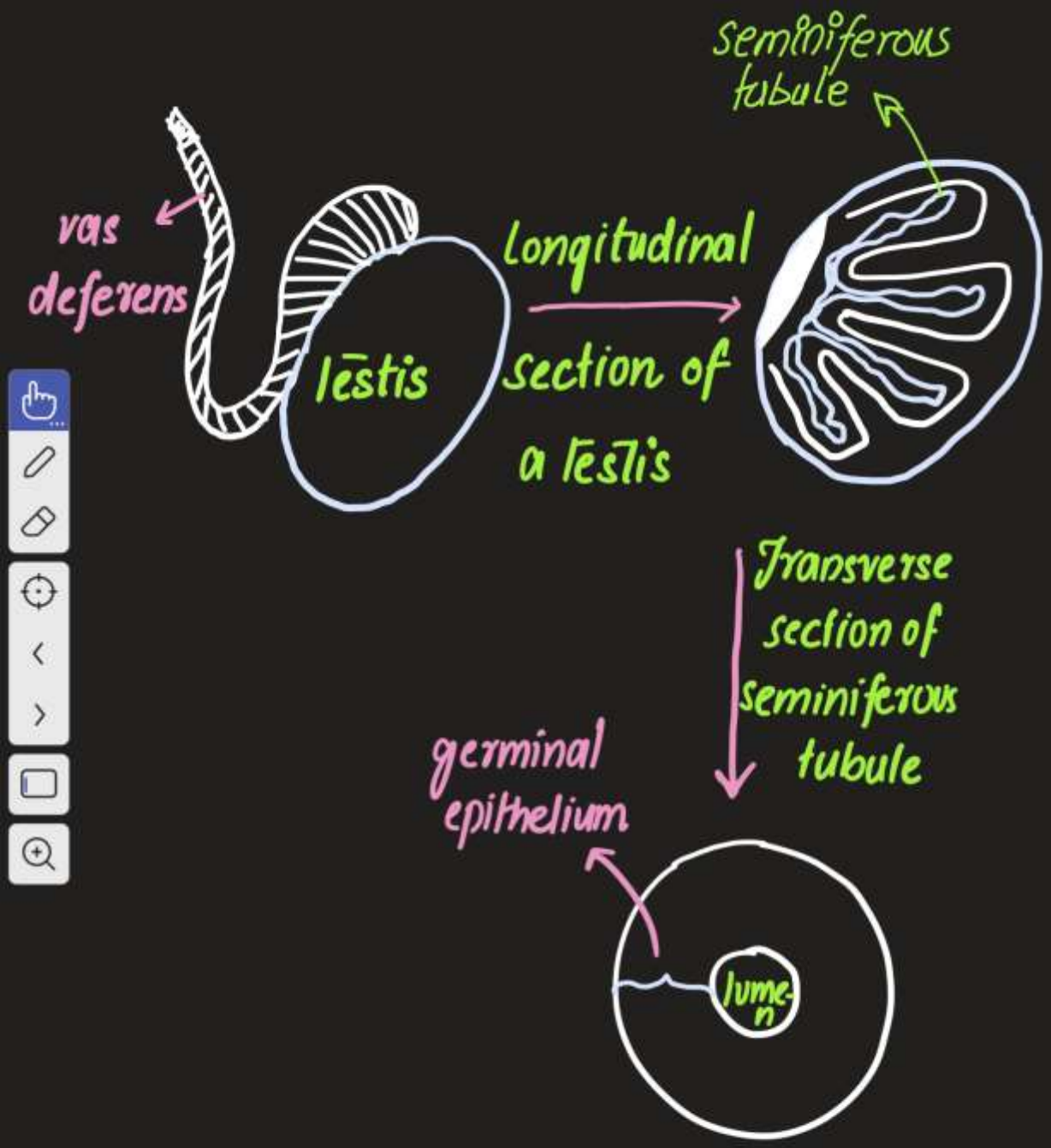
seminiferous tubule
vas deferens
testis
longitudinal section of a testis
transverse section of seminiferous tubule
germinal epithelium
lumen

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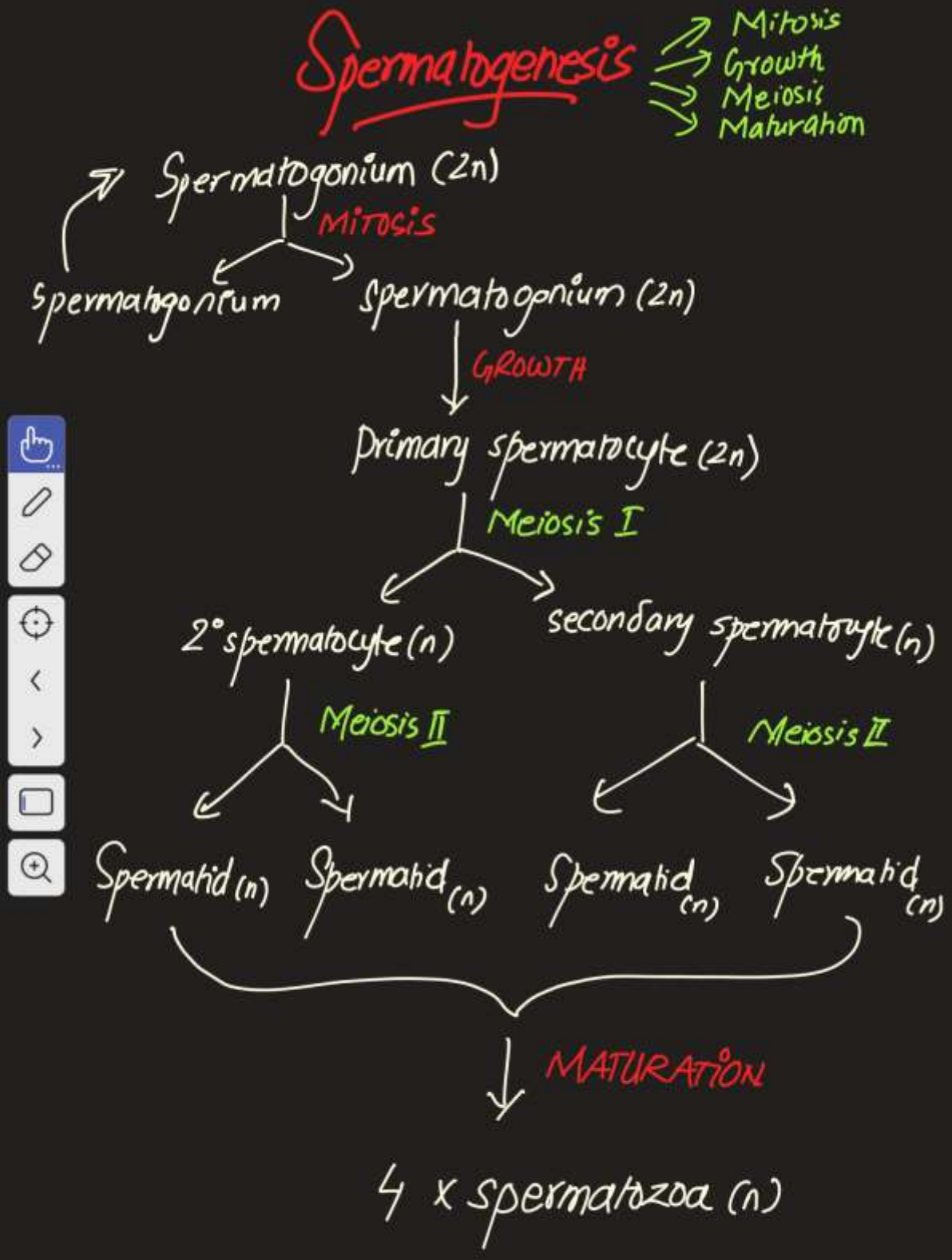
Spermatogenesis
Mitosis
Growth
Meiosis
Maturation
Spermatogonium (2n)
MITOSIS
Spermatogonium
Spermatogonium (2n)
GROWTH
Primary spermatocyte (2n)
Meiosis I
2° spermatocyte (n)
secondary spermatocyte (n)
Meiosis II
Meiosis II
Spermatid (n)
Spermatid (n)
Spermatid (n)
Spermatid (n)
MATURATION
4 x spermatozoa (n)

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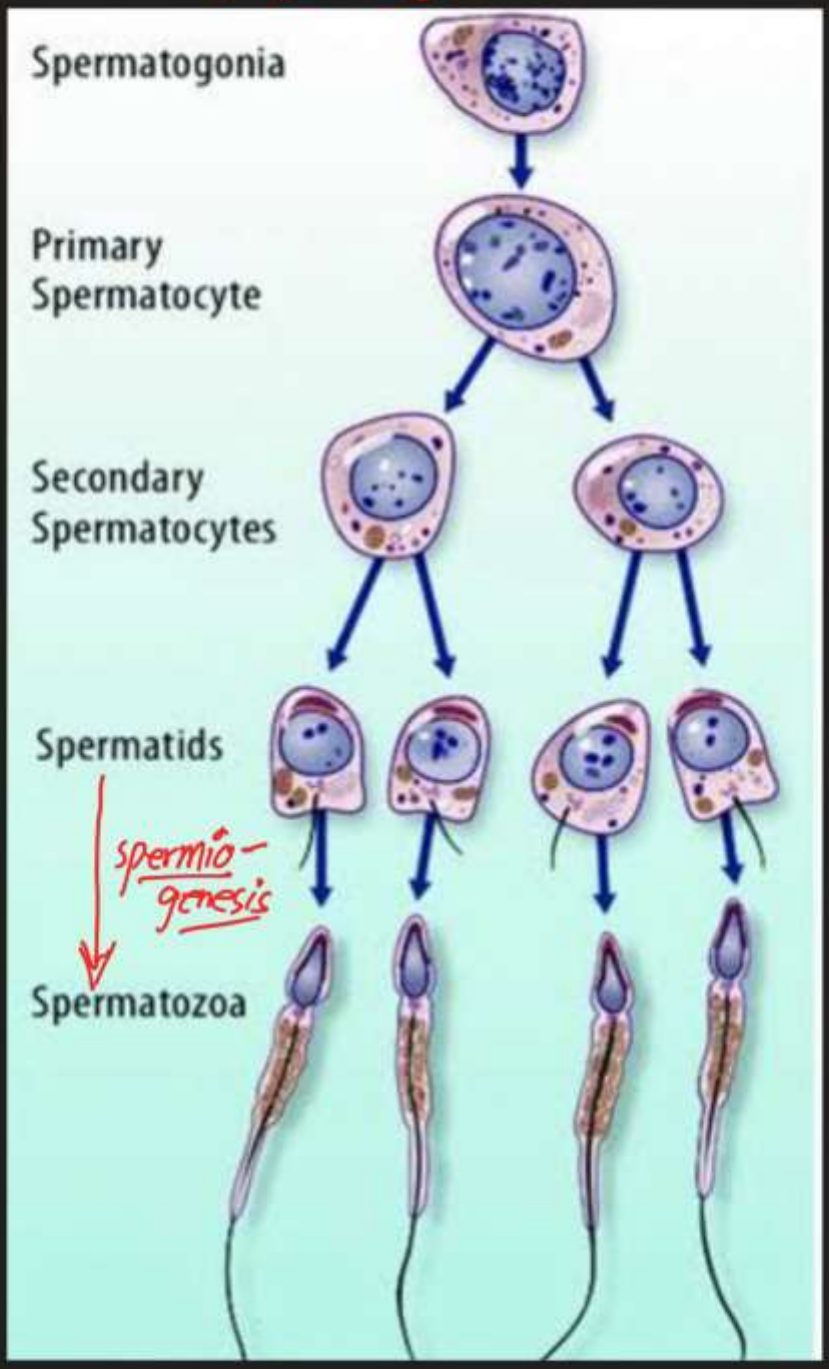
Spermatogenesis



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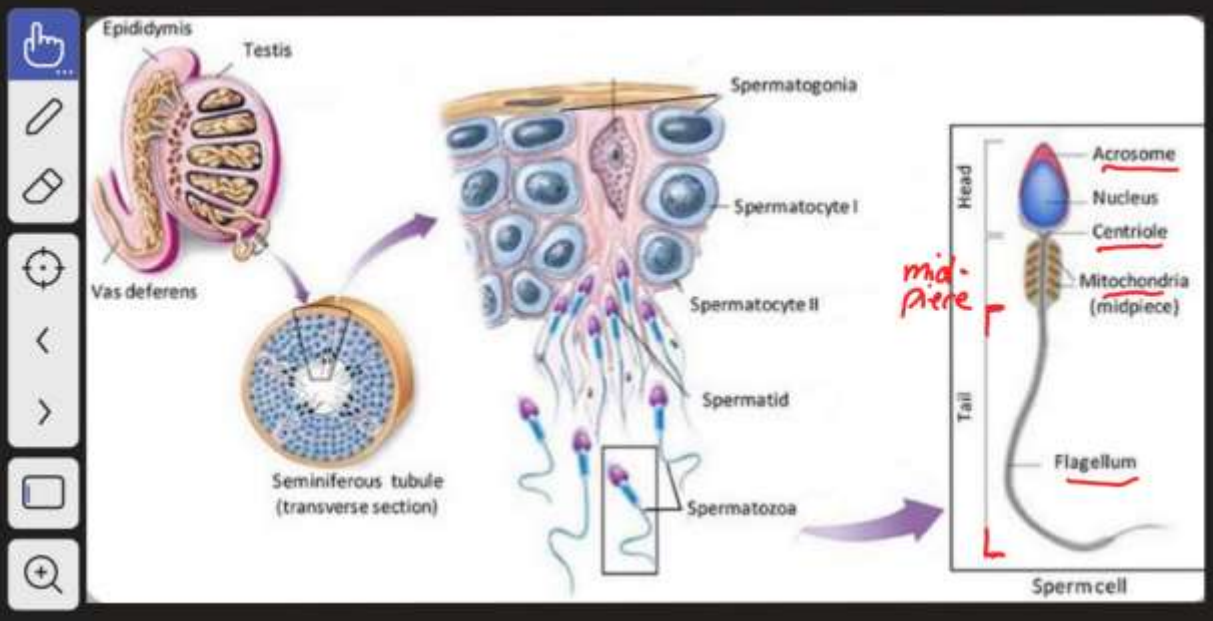
Epididymis
Testis
Vas deferens
Seminiferous tubule
(transverse section)
Spermatogonia
Spermatocyte I
Spermatocyte II
Spermatid
Spermatozoa
Head
Tail
Acrosome
Nucleus
Centriole
Mitochondria
(midpiece)
Flagellum
Sperm cell

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Stages of spermatogenesis

* Spermatogenesis occurs in four stages:

* Mitosis:

- Diploid spermatogonia present within the germinal epithelium divide repeatedly by mitosis to produce numerous diploid spermatogonia.

* Growth:

- Some diploid spermatogonia further grow to produce diploid primary spermatocytes.

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* Meiosis :

* Each primary spermatocyte divides by Meiosis I To produce Two haploid secondary spermatocytes. Each secondary spermatocyte divides by Meiosis II To produce Two haploid spermatids.

* Therefore one spermatogonium produces 4 haploid spermatids.

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Maturation :

- Each haploid spermatid undergoes a process of maturation to form a malure spermatozoon.
- This phase of maturation is known as spermiogenesis.
- During this phase each spermatid goes through a sequence of structural changes to form a motile spermatozoon. This involves :

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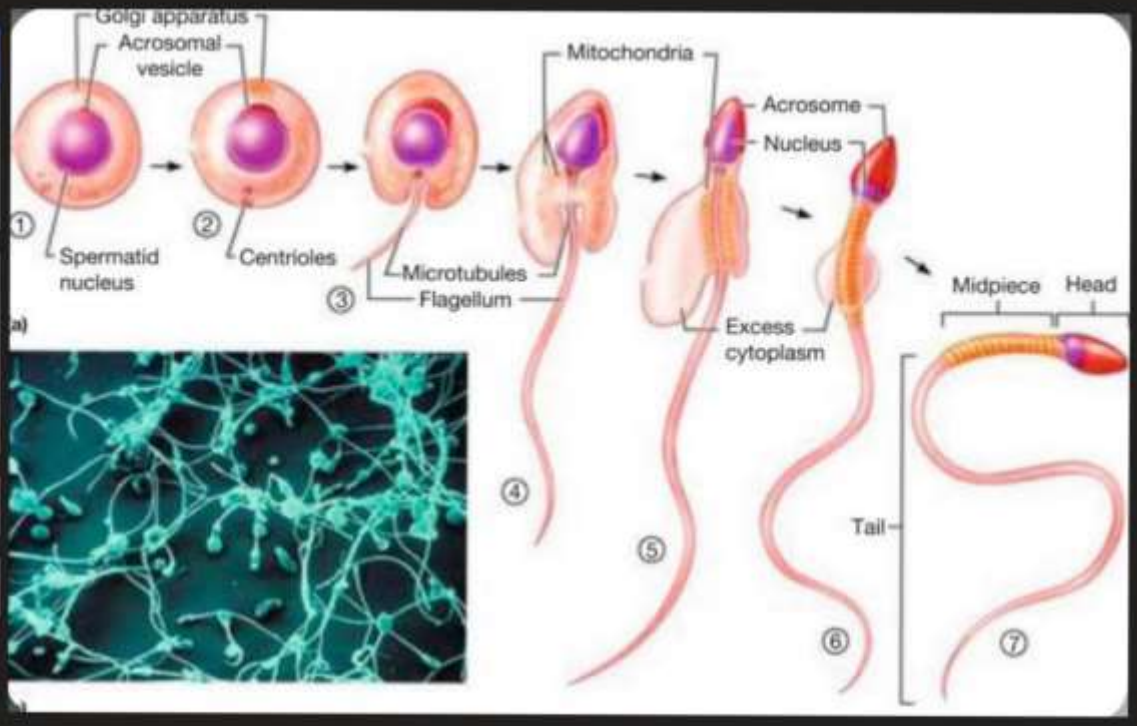
Golgi apparatus
Acrosomal vesicle
Mitochondria
Acrosome
Nucleus
Spermatid nucleus
Centrioles
Microtubules
Flagellum
Midpiece
Head
Excess cytoplasm
Tail
a)

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i) modification of the Golgi vesicles to form the acrosome which contains hydrolytic enzymes.

ii) The centrioles within the spermatids are responsible for forming the midpiece which contains the microtubules and the mitochondria.

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iii) The tail forms as a result of extension of the midpiece.

iv) The pear-shaped head of the spermatozoon results due to phagocytosis of excess cytoplasm of the spermatid.

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Oogenesis

* Oogenesis is a form of gametogenesis in females.

* The site of oogenesis are the female ovaries (germinal epithelium).

* Oogenesis has the following four stages:

1) Mitosis
2) Growth
3) Meiosis
4) Maturation

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* Oogenesis begins during The embryonic life.
* Oogenesis has long waiting stages. The phases of oogenesis can be sub-divided on The basis of The long waiting stages into:

a) Embryonic life
b) From puberty till menopause
c) At fertilisation.

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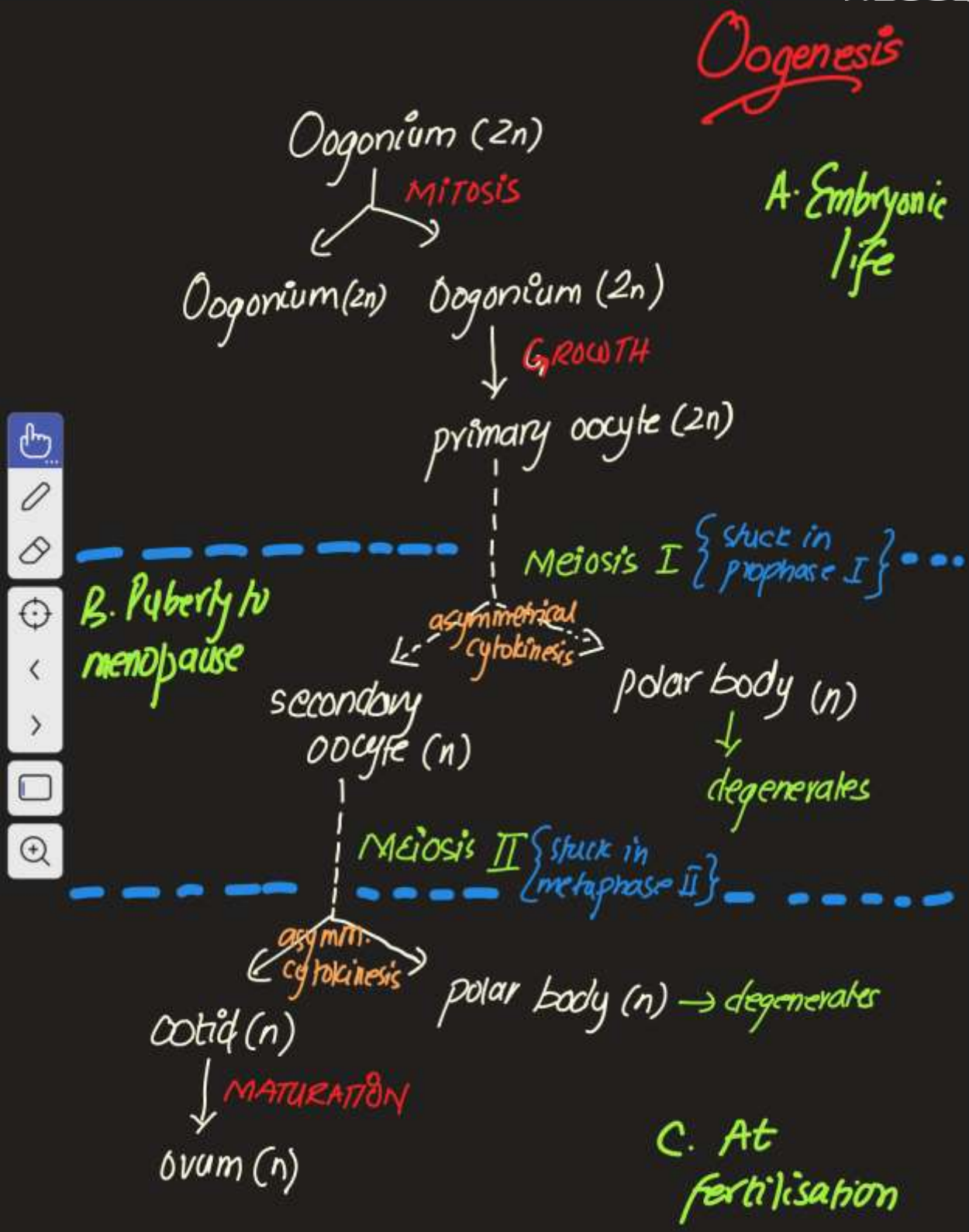
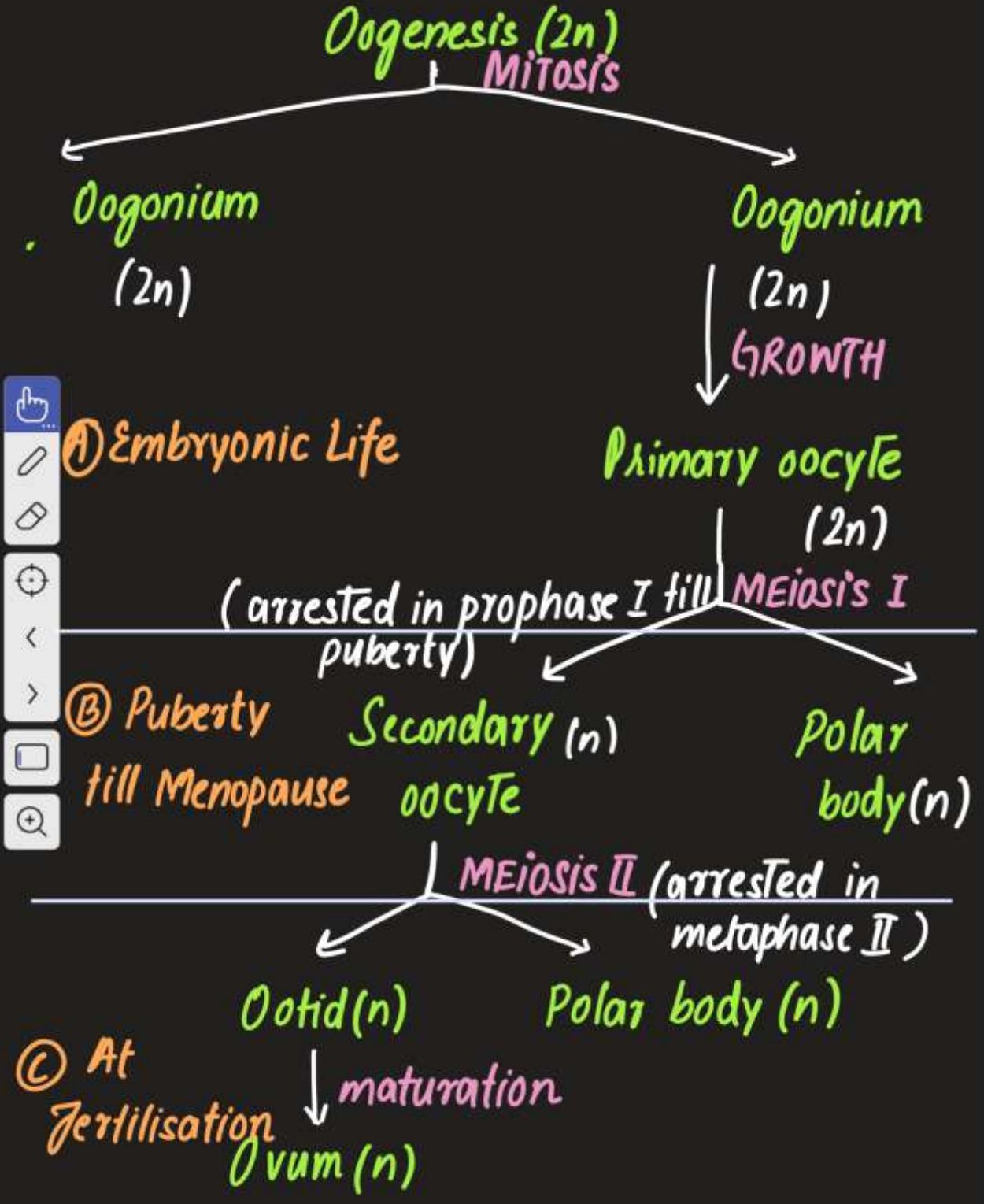

Oogenesis
Oogonium (2n)
MITOSIS
A. Embryonic life
Oogonium (2n)
Oogonium (2n)
GROWTH
primary oocyte (2n)
Meiosis I {stuck in prophase I}
B. Puberty to menopause
asymmetrical cytokinesis
secondary oocyte (n)
polar body (n)
degenerates
Meiosis II {stuck in metaphase II}
asymm. cytokinesis
polar body (n) → degenerates
ootid (n)
MATURATION
ovum (n)
C. At fertilisation


Oogenesis (2n)
Mitosis
Oogonium
(2n)
Oogonium
(2n)
GROWTH
A Embryonic Life
Primary oocyte
(2n)
(arrested in prophase I till puberty)
Meiosis I
B Puberty till Menopause
Secondary (n) oocyte
Polar body (n)
Meiosis II (arrested in metaphase II)
Ootid (n)
Polar body (n)
C At Fertilisation
maturation
Ovum (n)

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Embryonic Life:

* Diploid oogonia within the germinal epithelium divide repeatedly by mitosis.
* Some diploid oogonia pass through a growth phase to form diploid primary oocytes.
* Each primary oocyte starts Meiosis I and remains suspended in prophase I till puberty.
* At birth a female has approximately two million primary oocytes.
* Following birth there is no further production of primary oocytes.

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From Puberty till Menopause

* A female undergoes monthly cyclical changes within the ovary following menarche.

* Each month a primary oocyte completes Meiosis I to form a secondary oocyte & a polar body.

* The polar body degenerates to get rid of excess genetic material.

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* The secondary oocyte contains most of the cytoplasm due to asymmetrical cytokinesis.

* The secondary oocyte starts Meiosis II but remains suspended at Metaphase II.

* The secondary oocyte is released during ovulation.

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At fertilisation

* The secondary oocyte completes Meiosis II if fertilisation occurs to form an ootid and the second polar body.
* The ootid formed rapidly completes its maturation phase to form a mature ovum.
* The nucleus of the ovum fuses with the nucleus of a sperm cell to form a diploid zygote.

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* NOTE: Asymmetrical cytokinesis occurs twice (because polar bodies are formed twice) to ensure that the mature ovum contains all the cytoplasm and organelles within it.

* The formation of polar bodies gets rid of excess genetic material.

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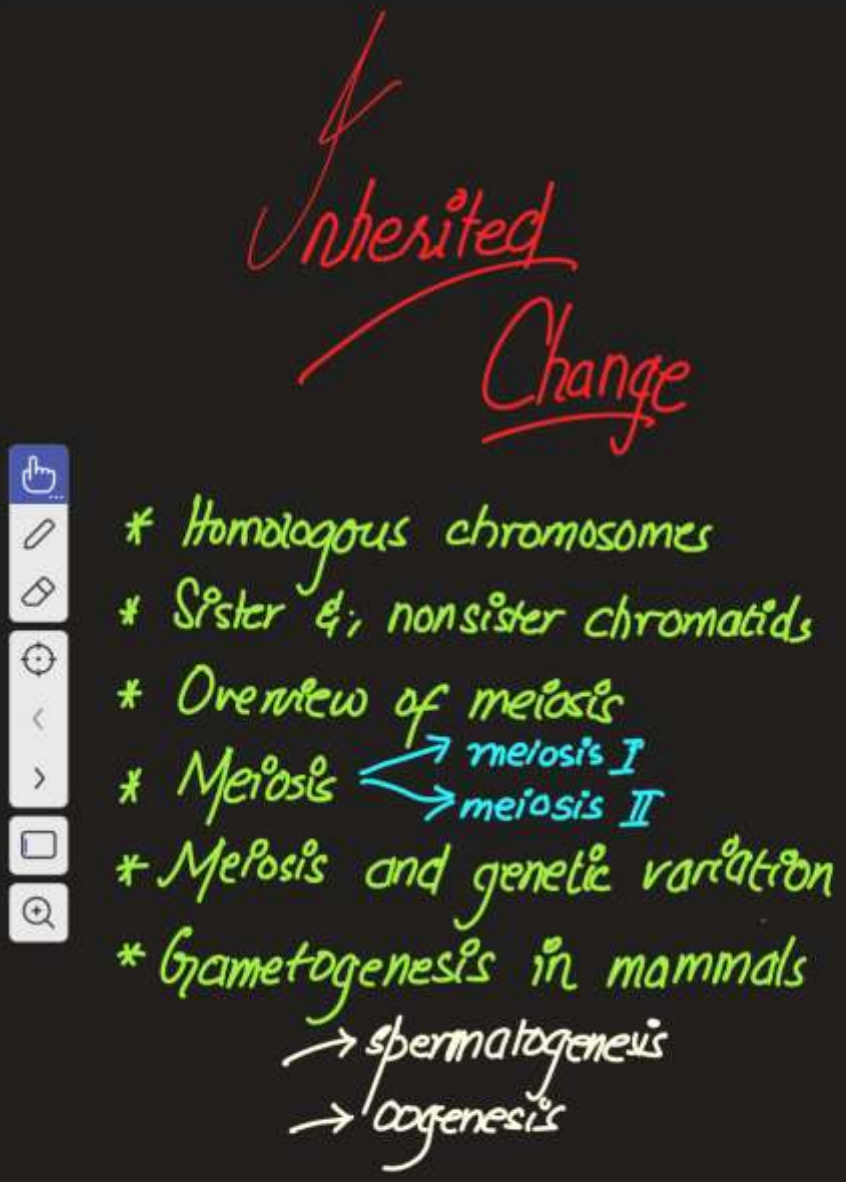
Inherited Change



With
Mohammad Hussham Arshad, MD

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ADVANCED LEVEL BIOLOGY 9700

UNIT 16: Inherited Change

Learning Objectives:

- **Comparing spermatogenesis & oogenesis**
- **Gametogenesis in plants**
- **Comparing pollen grain formation & embryo sac formation**
- **Comparing gametogenesis in mammals & plants**

Video Lecture 4 Slides

Mohammad Hussham Arshad, MD
Biology Department

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Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis
* Meiosis → meiosis I, meiosis II
* Meiosis and genetic variation
* Gametogenesis in mammals
  - → spermatogenesis
  - → oogenesis

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Comparing spermatogenesis and oogenesis

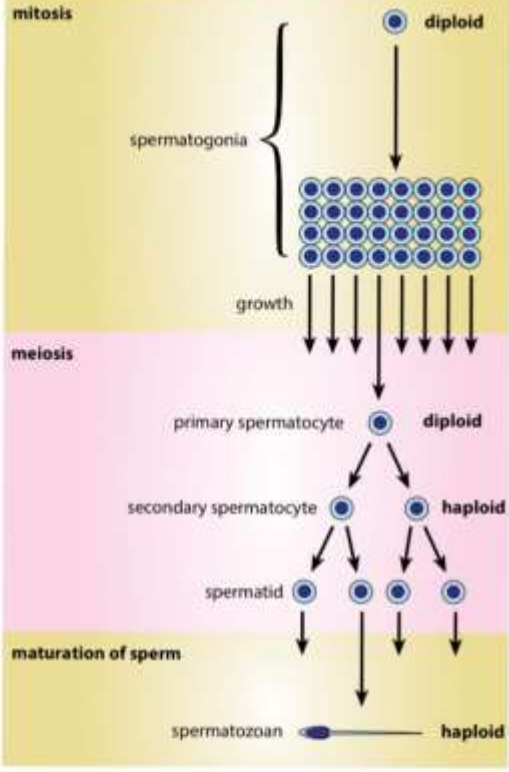

Figure 16.10 Spermatogenesis in humans.

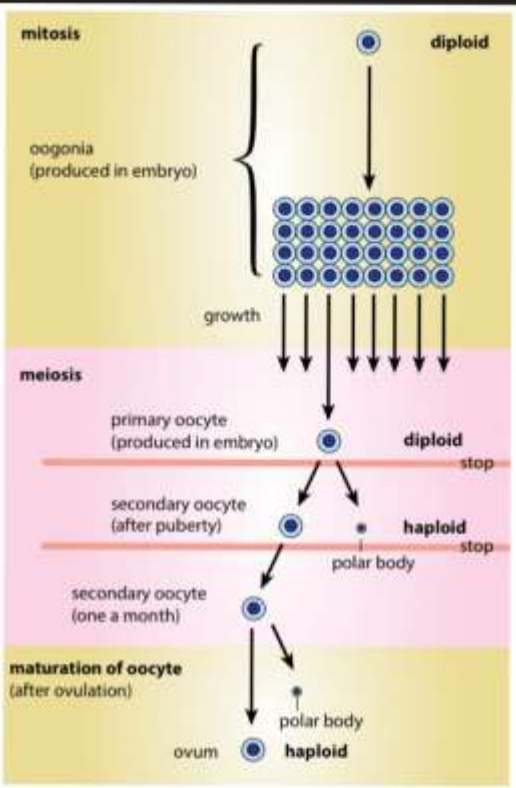

Figure 16.11 Oogenesis in humans.

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Q1: Differentiate between spermatogenesis & oogenesis?

Ans:

Spermatogenesis	Oogenesis
• Four haploid spermatozoa are formed from one diploid spermatogonium.	• One haploid ovum forms from one diploid oogonium.
• Does not involve polar bodies.	• Involves production of polar bodies.
• Symmetrical cytokinesis	• Asymmetrical cytokinesis.

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Spermatogenesis	Oogenesis
• Spermatogenesis begins following puberty.	• Oogenesis begins during embryonic life.
• Continuous process with no waiting stages	• Interrupted by long waiting stages.
• Spermatogenesis occurs within the testis	• Occurs within the ovary.

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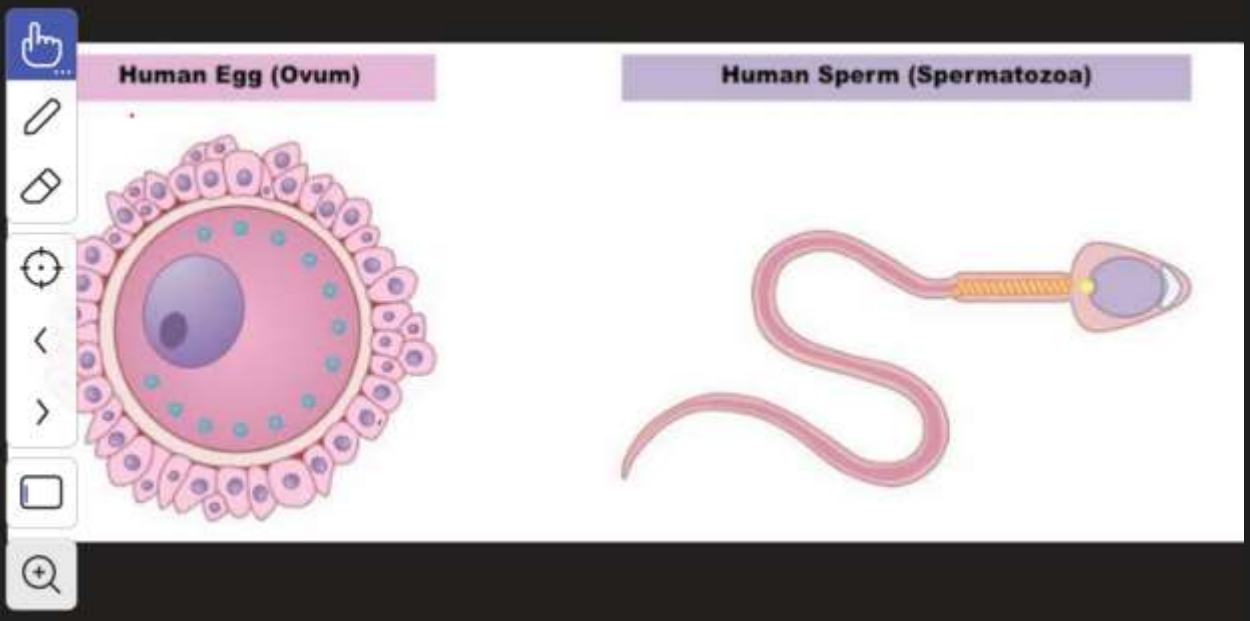
Comparing an ovum with a spermatozoa



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Q2: Compare and contrast an ovum with a spermatozoa?

Ans: Similarities:

i) Both have 1 set of chromosomes and are therefore haploid.

ii) Both are produced as a result of gameto-genesis by germ cells.

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Differences :

Sperm Cell	Ovum
i) Spermatozoa are motile	i) Ova is immotile
ii) Smaller in size	ii) Larger in size
iii) Has comparatively less cytoplasm.	iii) Has comparatively large volume of cytoplasm
iv) Contain an acrosome with hydrolytic enzymes.	iv) Does not contain an acrosome
v) Has a tail.	v) Ova are devoid of a tail.

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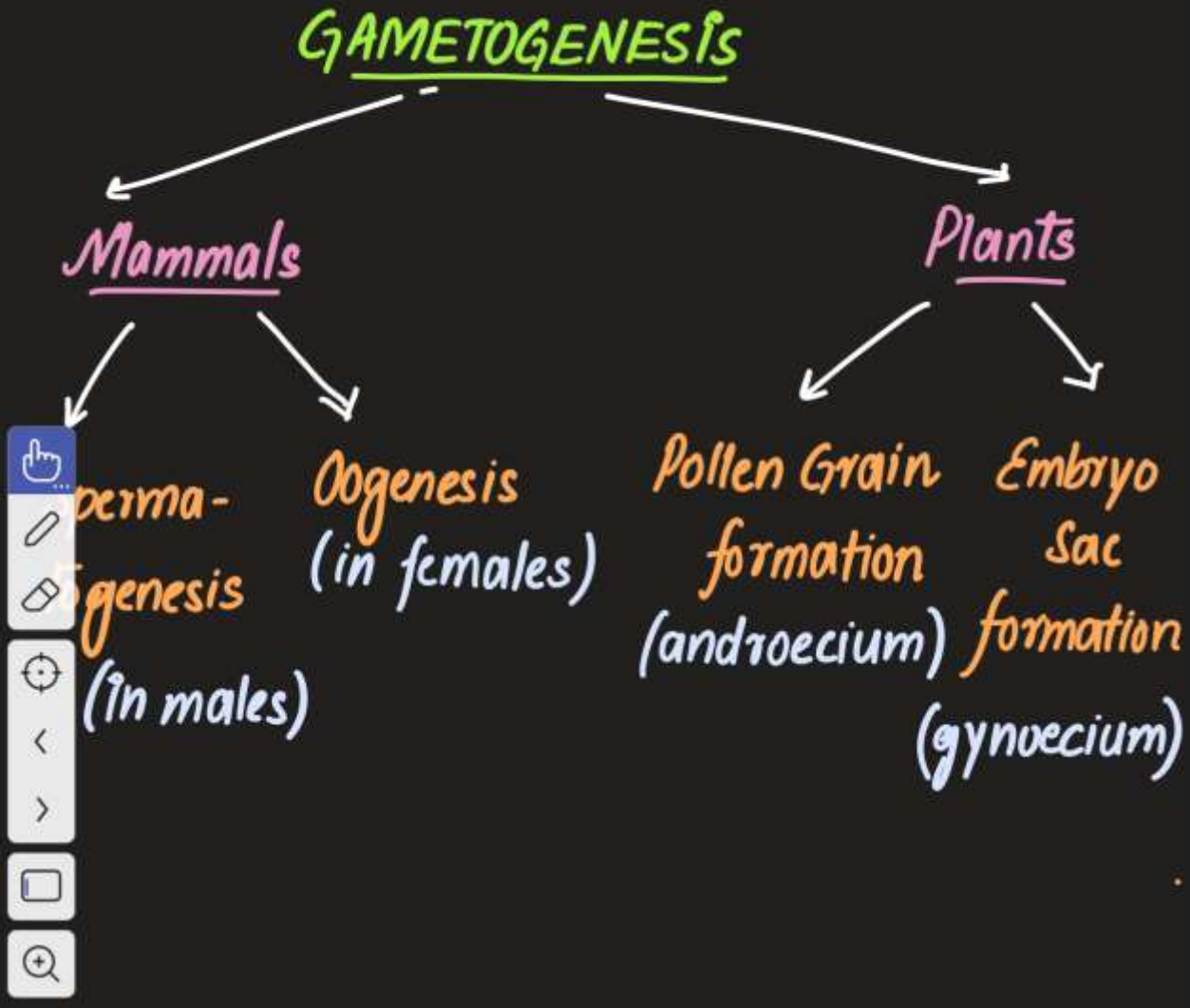
GAMETOGENESIS
Mammals
Plants
perma-
genesis
(in males)
Oogenesis
(in females)
Pollen Grain
formation
(androecium)
Embryo
Sac
formation
(gynoecium)

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Gametogenesis in flowering plants

* Gametogenesis in flowering plants leads to formation of mature pollen grains and mature embryo sac.

Pollen grain formation occurs in the male parts of the flowering plant collectively termed as **stamen** or **androecium**.

* Parts of androecium include:

a) Anthers

b) Filament

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* Embryo sac formation occurs in the female parts of the flowering plants collectively termed as the carpel or the gynoecium.

* Parts of the gynoecium include:

a) Stigma
b) Style
c) Ovary
d) Ovule

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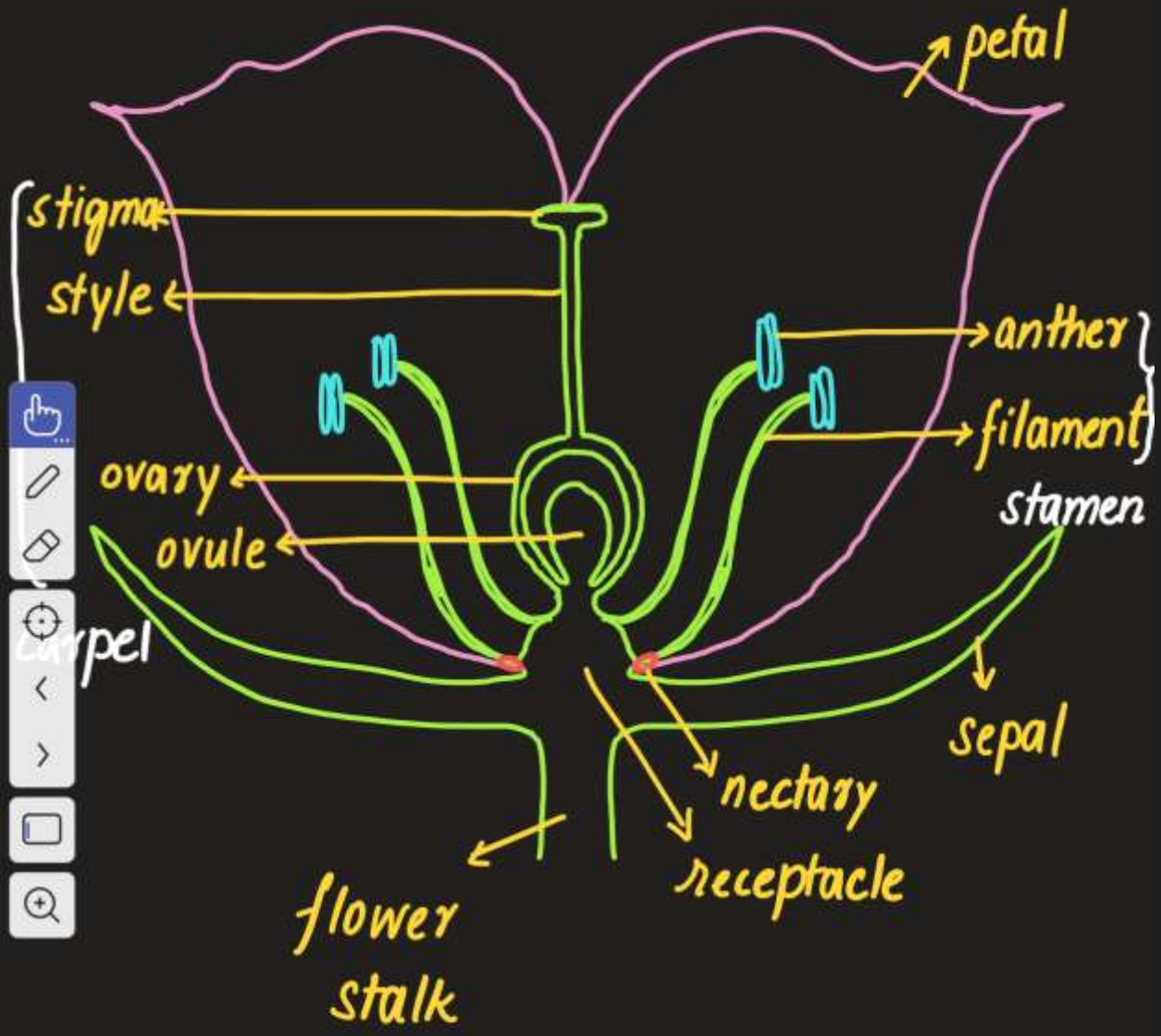
petal
stigma
style
anther
filament
stamen
ovary
ovule
carpel
sepal
nectary
receptacle
flower
stalk

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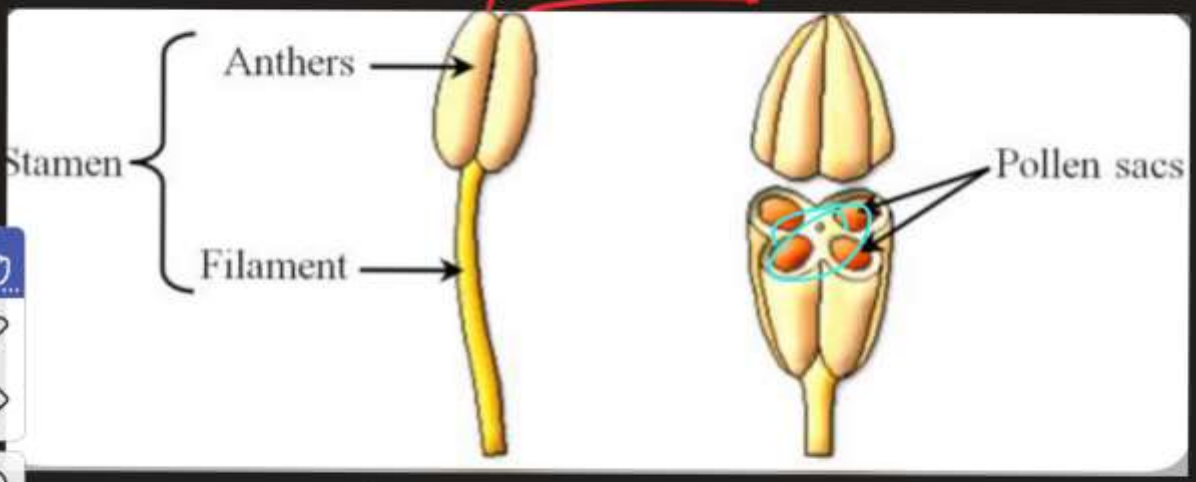
Pollen grain formation



Pollen mother cell (2n)

↓ meiosis

four haploid cells (n)

↓ mitosis without cytokinesis

generative n.

tube n.

immature pollen grains

1. Inner layer (intine)
2. Outer layer (exine)

↓

mature pollen grains (n)

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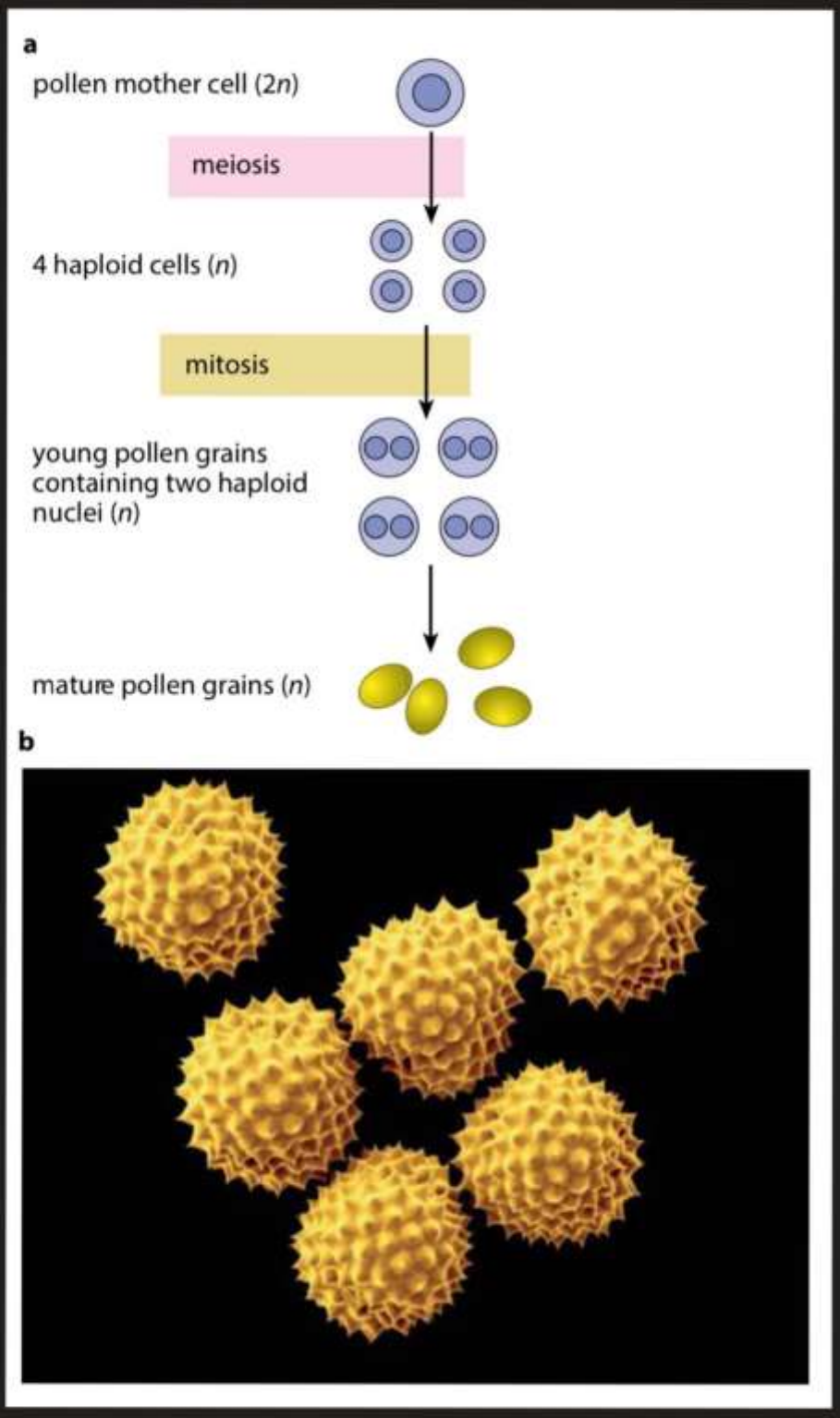
a
pollen mother cell (2n)
meiosis
4 haploid cells (n)
mitosis
young pollen grains containing two haploid nuclei (n)
mature pollen grains (n)
b

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Pollen grain formation

* Pollen grains are formed in pollen sacs inside The anthers.

* Pollen sacs contain diploid pollen mother cells. A diploid pollen mother cell divides by meiosis To produce four haploid cells.

* Each haploid cell undergoes mitosis without cytokinesis To produce the cell with Two haploid nuclei. These nuclei are the gametic nuclei.

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* One of the nucleus is termed as the tube nucleus and the other as the generative nucleus.

* The cell with 2 haploid nuclei forms a protective coat with 2 layers, an inner intine and an outer exine. This mature cell is termed as a pollen grain.

* One diploid pollen mother cell therefore produces four mature pollen grains.

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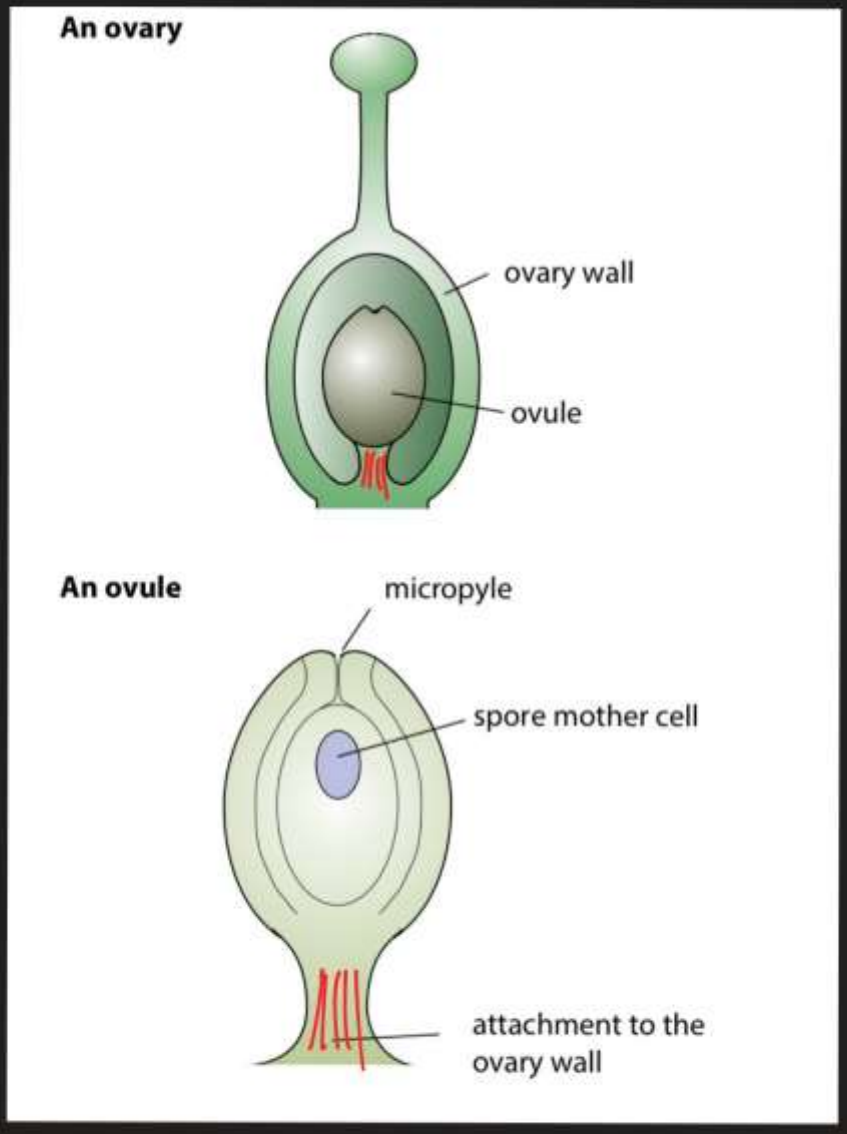
Embryo Sac formation



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Embryo Sac formation

spore mother cell (2n)

↓ meiosis

4 × haploid cells (n)

{ 3 are crushed by the micropyle }

↓

1 × haploid cell (n)

↓

↓ mitosis × 3 WITHOUT cytokinesis

↓

mature embryo sac

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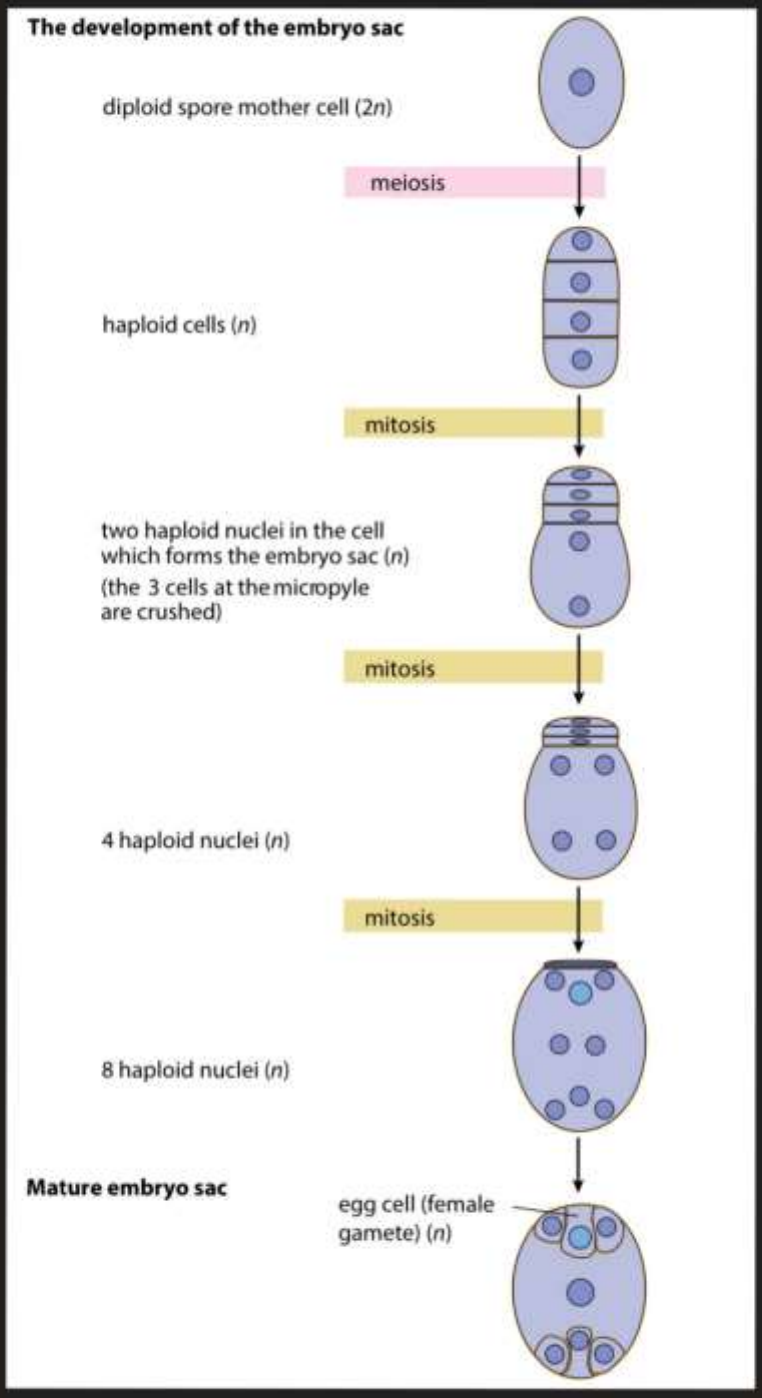
The development of the embryo sac
diploid spore mother cell (2n)
meiosis
haploid cells (n)
mitosis
two haploid nuclei in the cell which forms the embryo sac (n)
(the 3 cells at the micropyle are crushed)
mitosis
4 haploid nuclei (n)
mitosis
8 haploid nuclei (n)
Mature embryo sac
egg cell (female gamete) (n)

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Embryo Sac Formation

* Embryo sac formation occurs in the ovule.
* A diploid spore mother cell within the ovule divides by meiosis to form four haploid cells.
* Three out of these four cells are crushed by the micropyle.
* The remaining haploid cell undergoes three mitotic divisions to form a mature embryo sac.

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* A mature embryo sac has eight haploid nuclei, one of which is a gametic nuclei.
* One diploid spore mother cell thereafter produces one mature embryo sac.
* Fertilisation occurs when a male gamete within a pollen grain fuses with the ovule to form a diploid zygote. This zygote divides repeatedly by mitosis to form the embryo.

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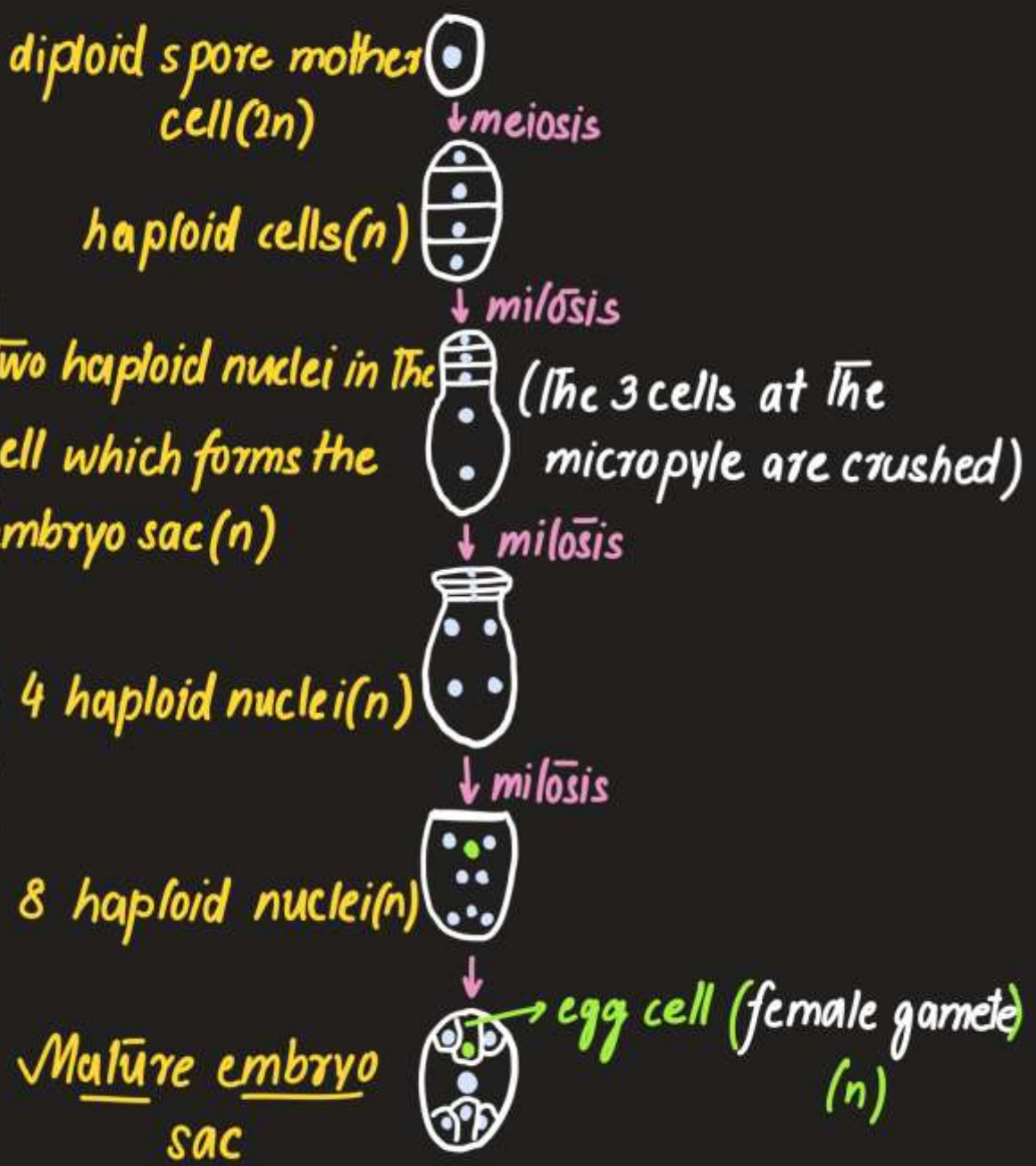
The development of the embryo sac



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Comparing pollen grain and embryo sac formation

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Q: Compare and contrast pollen grain formation and embryo sac formation.

Similarities:

* Both involve mitosis and meiosis (before mitosis)
* Both involve The formation of gametes via mitosis of haploid nuclei.
* Both involve mitosis without cytokinesis.

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Differences:

Pollen Grain Formation	Embryo Sac Formation
1) Pollen grains are formed from diploid pollen mother cells.	Embryo sac is formed from diploid spore mother cells.
2) One pollen mother cell produces 4 mature pollen grains.	One diploid spore mother cell produces 1 mature embryo sac.
3) Haploid nucleus within the pollen grain divides once by mitosis.	Haploid nucleus within the embryo sac divides thrice by mitosis.

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Pollen Grain Formation	Embryo Sac Formation
4) A mature pollen grain has 2 haploid nuclei.	A mature embryo sac has 8 haploid nuclei.

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Compare gametogenesis in mammals and plants

* Spermatogenesis Vs Pollen grain formation

Similarities
- meiosis produces 4 cells
- Chromosome number halves
- produce sentially different cells

Differences:

Spermatogenesis	Pollen grain formation
1. Mature sperm has one haploid nucleus	1. Mature pollen grain has two haploid nuclei
2. Mitosis occurs before meiosis	2. Meiosis occurs before mitosis
3. Primary spermatocyte undergoes meiosis	3. Pollen mother cell undergoes meiosis

* Oogenesis Vs Embryo sac formation

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Questions

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3 Fig. 3.1 shows the structure of part of a seminiferous tubule of a human testis.

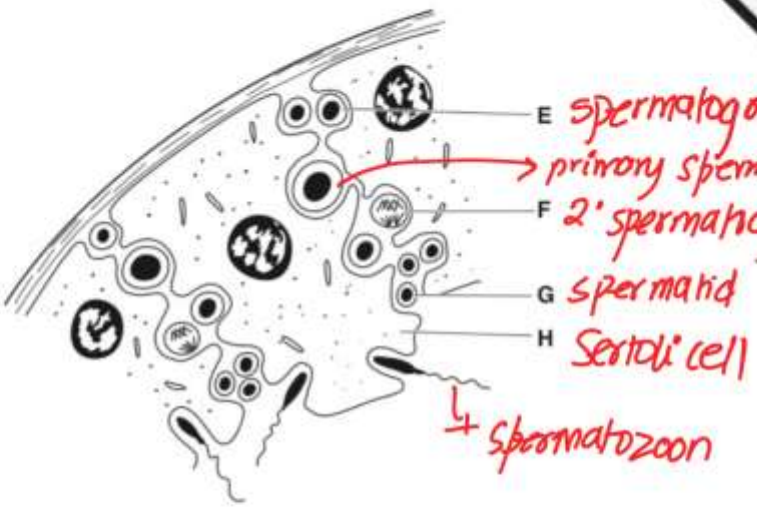

Fig. 3.1

(a) Name cells **E** to **H**.

E spermatogonium

F secondary spermatocyte

G Spermatid

H Sertoli cell [4]

(b) Describe how cell **F** is produced from cell **E** in the process of spermatogenesis.

* Spermatogonium is a diploid germ cell
* undergoes a growth phase (increase in size)
* to form a primary spermatocyte
* primary spermatocyte undergoes meiosis I
* to form haploid secondary spermatocyte.

[4]

[Total: 8]

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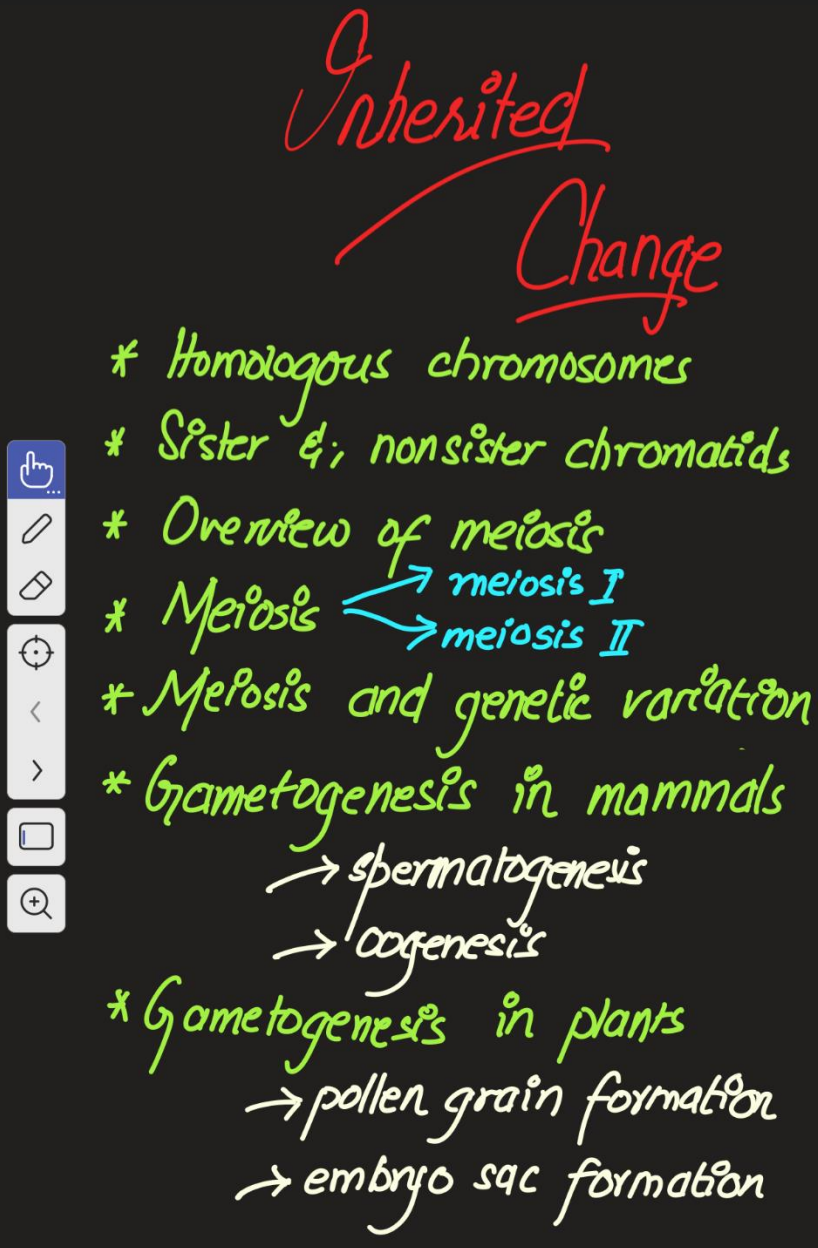
Inherited Change



With
Mohammad Hussham Arshad, MD

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ADVANCED LEVEL BIOLOGY 9700

UNIT 16: Inherited Change

Learning Objectives:

- **Monohybrid inheritance I**

Video Lecture 5 Slides

Mohammad Hussham Arshad, MD
Biology Department

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Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis
* Meiosis → meiosis I, meiosis II
* Meiosis and genetic variation
* Gametogenesis in mammals
  - → spermatogenesis
  - → oogenesis
* Gametogenesis in plants
  - → pollen grain formation
  - → embryo sac formation

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Questions

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Q

4 **(a)** Fig. 4.1 shows the stages in spermatogenesis in a mammal.

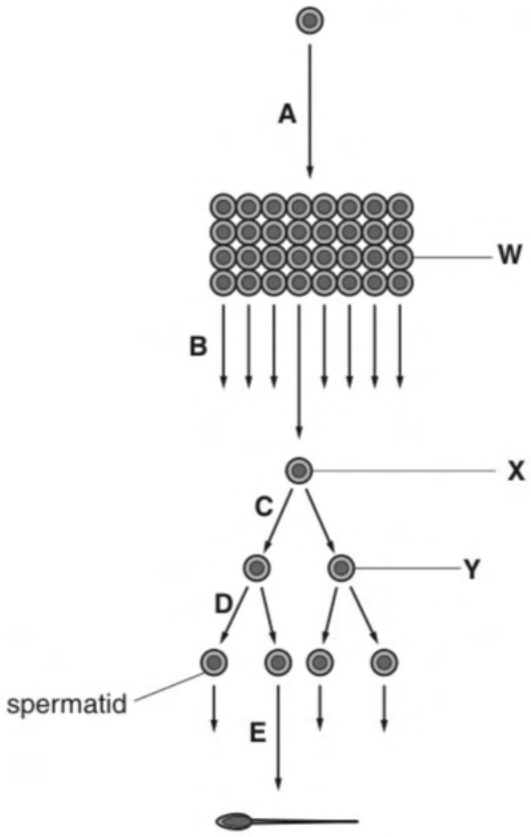

Fig. 4.1

(i) State the letter(s) of the arrow or arrows that represent mitosis.

A ..... [1]

(ii) Name the cells **W**, **X** and **Y**.

W Spermatogonium

X primary spermatocyte

Y secondary spermatocyte ..... [3]

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(c) In all animals so far studied, the production of fully functional sperm is sensitive to temperature.

In the nematode worm, *Caenorhabditis elegans*, spermatogenesis takes place in a similar way to mammals. Two proteins known as argonaute proteins are important in the development of sperm. They are coded for by the genes *alg-3* and *alg-4*.

Table 4.1 shows the effect of mutations in one or both of these genes on the fertility of male worms, at temperatures of 20 °C and 25 °C.

Fertility was measured as the mean number of offspring produced when the male worms mated with normal females.

Table 4.1

male worms	mean number of offspring produced	
	at 20 °C	at 25 °C
normal at both gene loci	280	150
mutation in *alg-3* only	125	95
mutation in *alg-4* only	220	85
mutations in **both** *alg-3* **and** *alg-4*	90	0

(i) Describe the effect of increased temperature on the fertility of normal male worms.

* fertility of male worms decreases at higher temp.
* At 20°C, mean no. of offsprings produced = 280
At 25°C, " " " " " " " = 150

[2]

(ii) Compare the effect of increased temperature on the fertility of *alg-3* mutant male worms with the effect on fertility of *alg-4* mutant male worms.

* male worms with mutation in alg-4 have a mae significant decrease in fertility at higher temp.
* mean no. of offsprings dearease from 220 to 85 in alg-4 mutants whereas they dec. from 125 to 95 in alg-3 mutants [2]

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Q

4 **(a)** Meiosis has an important role in sexual reproduction. Meiosis occurs during gametogenesis in humans and during the formation of pollen grains and embryo sacs in plants.

(i) Complete Table 4.1 to compare meiosis with mitosis.

For both columns, put a tick (✓) in the box if the statement is correct and put a cross (✗) in the box if the statement is incorrect.

Each box must contain either a tick or a cross.

Table 4.1

statement	meiosis	mitosis
chromosome number is maintained	✗	✓
homologous chromosomes pair up	✓	✗
sister chromatids separate	✓	✓
occurs in prokaryotes	✗	✗

[2]

(ii) Compare the role of meiosis in gametogenesis to produce sperm cells in humans with the role of meiosis in gametogenesis in producing pollen grains in flowering plants.

* a sperm cell has one haploid nucleus and a pollen grain has two haploid nuclei
* primary spermatocyte undergoes meiosis in spermatogenesis whereas a pollen mother cell undergoes meiosis in pollen grain formation
* Spermatogenesis involves mitosis before meiosis whereas pollen grain formation involves meiosis before mitosis

[4]

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Genetic Inheritance

Monohybrid Inheritance

Types

* Dominant vs Recessive
* Codominance
* Multiple alleles
* Test cross
* Sex-linkage
* Lethal alleles

Dihybrid Inheritance

- without crossing over
- with crossing over

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Monohybrid Inheritance

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MONOHYBRID INHERITANCE

* Monohybrid inheritance refers to the inheritance of one gene.

* We will be studying the following genetic crosses as types of monohybrid inheritance:

a) Dominant & Recessive Alleles

b) Codominant alleles (codominance)

c) Multiple Alleles

d) Test Cross

e) Sex linked Alleles

f) Lethal Alleles

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Dominant & Recessive alleles

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(A) Dominant & Recessive alleles

(1) Gregor Mendel discovered the fundamental laws of inheritance through his ground-breaking work on pea plants.

(2) He studied the inheritance of 7 traits in pea plants.

(3) Three of the 7 traits he studied are shown in Figures 1a, 1b and 1c.

(4) Through his work, he deduced that a gene comes in pairs (alleles) → one from each parent.

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(5) An allele is said to be **dominant** if it expresses itself in the phenotype in a homozygote and a heterozygote.

(6) An allele is said to be **recessive** if it expresses itself in the phenotype only in a homozygote.

(7) **Genotype** refers to genetic constitution of an organism.

(8) **Phenotype** refers to the physical detectable characteristic of an organism which results due to interaction b/w

the organism's genotype & it's environment.

⑨ An organism is said to be homozygous if it carries identical alleles of a gene.

⑩ An organism is said to be heterozygous if it carries different alleles of a gene.

⑪ Fig 1a shows a cross between pure breeding tall and short plants. Pure breeding strains are homozygous for a particular allele. When self-fertilised, pure-breeding strains always produce

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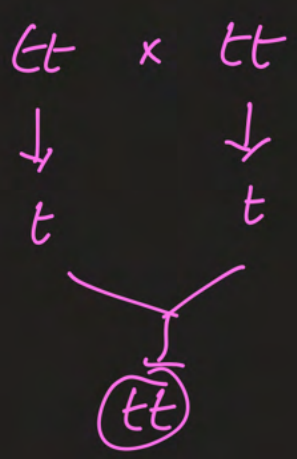
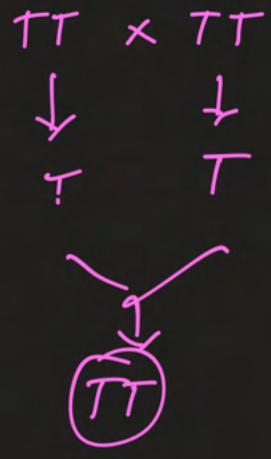
offsprings having the same genotype and phenotype.

Pure breeding strains

pure breeding short plant → self-fertilise → all offsprings are short

pure breeding tall plant → self-fertilise → all offsprings are tall





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Mendel experiments

Height of the pea plant

pure breeding parents: Tall x Short

F_1 generation: Tall

F_2 generation: Tall : Short = 3 : 1

Shape of the seeds

pure breeding parents: Round x Wrinkled

F_1 generation: Round seeds

F_2 generation: Round seeds : Wrinkled seeds = 3 : 1

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Colour of the seeds

F_1 – first filial generation

F_2 – second filial generation

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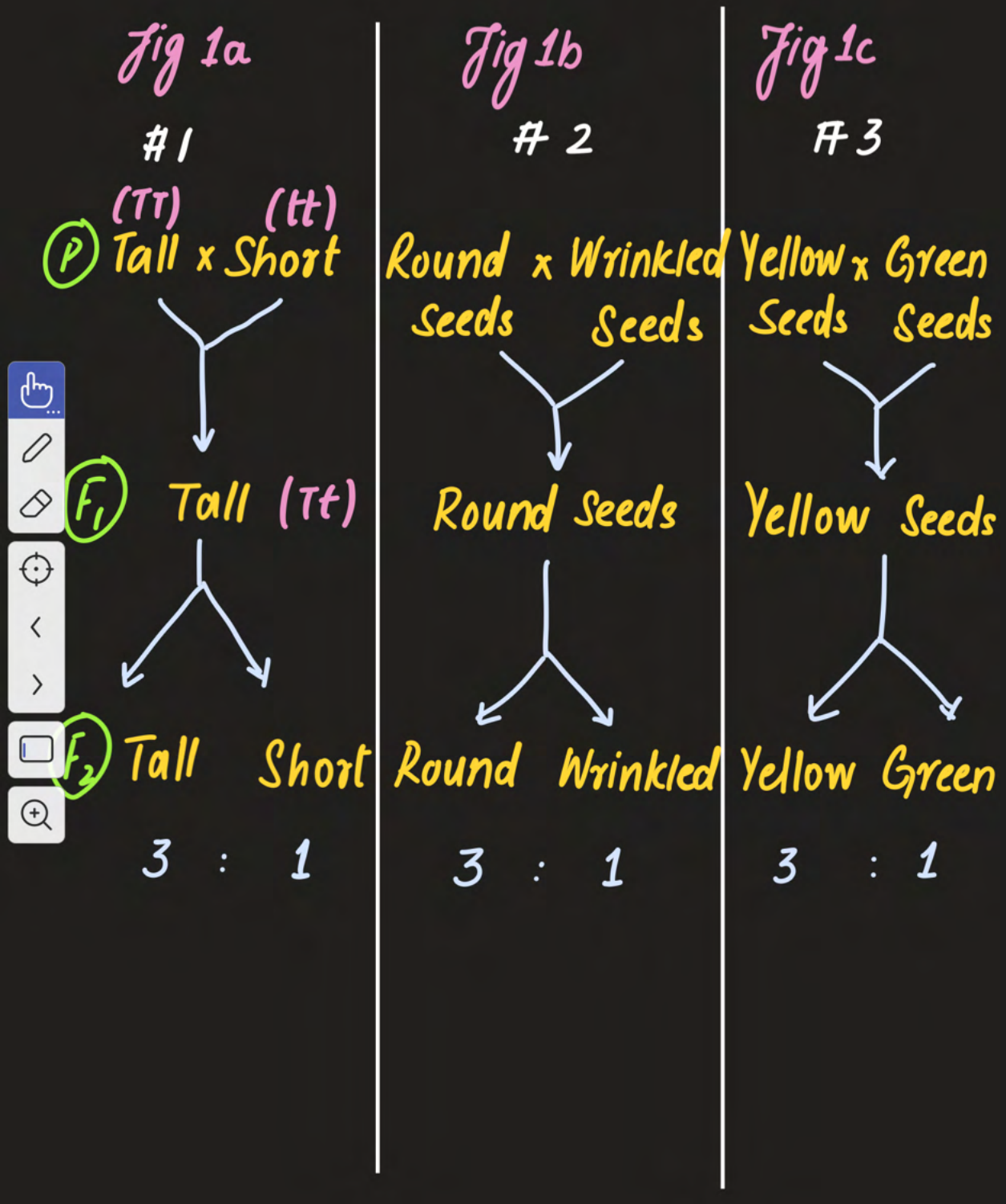
Fig 1a
#1
(TT) (tt)
P
Tall x Short
F1
Tall (Tt)
F2
Tall Short
3 : 1
Fig 1b
2
Round x Wrinkled
Seeds Seeds
Round Seeds
Round Wrinkled
3 : 1
Fig 1c
3
Yellow x Green
Seeds Seeds
Yellow Seeds
Yellow Green
3 : 1

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(11) Cross between pure breeding tall and short plant produces the F_1 generation.

(12) Cross b/w two F_1 organisms produces the F_2 generation (figure 2)

(13) The F_1 generation refers to the offsprings that result from a cross between homozygous dominant organisms and homozygous recessive organisms. All the offsprings in the F_1 generation are heterozygous.

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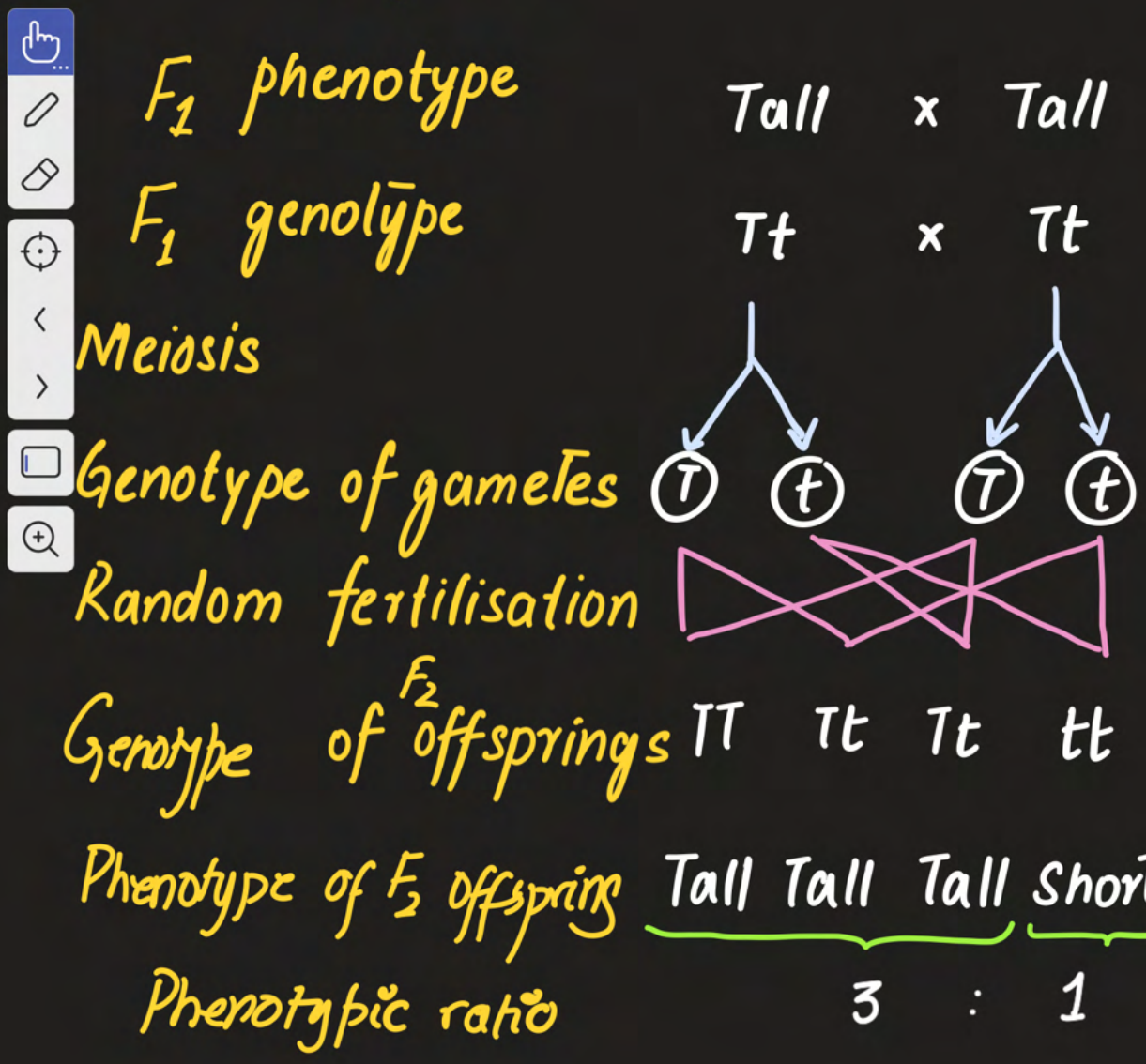
(14) F_2 generation refers to the offsprings that result from a cross b/w two F_1 organisms.

Figure 2



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Punnett Square

male → ♂
female → ♀

♀ \ ♂	T 1/2	t 1/2
T 1/2	TT 1/4	Tt 1/4
t 1/2	Tt 1/4	tt 1/4

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Q: Why did Mendel choose pea plants for his experiments?

Ans: (a) Pea plants grow really fast.

(b) Pea plants can undergo self-pollination and cross-pollination.

(c) Pea plants have sharply defined traits with two variants.

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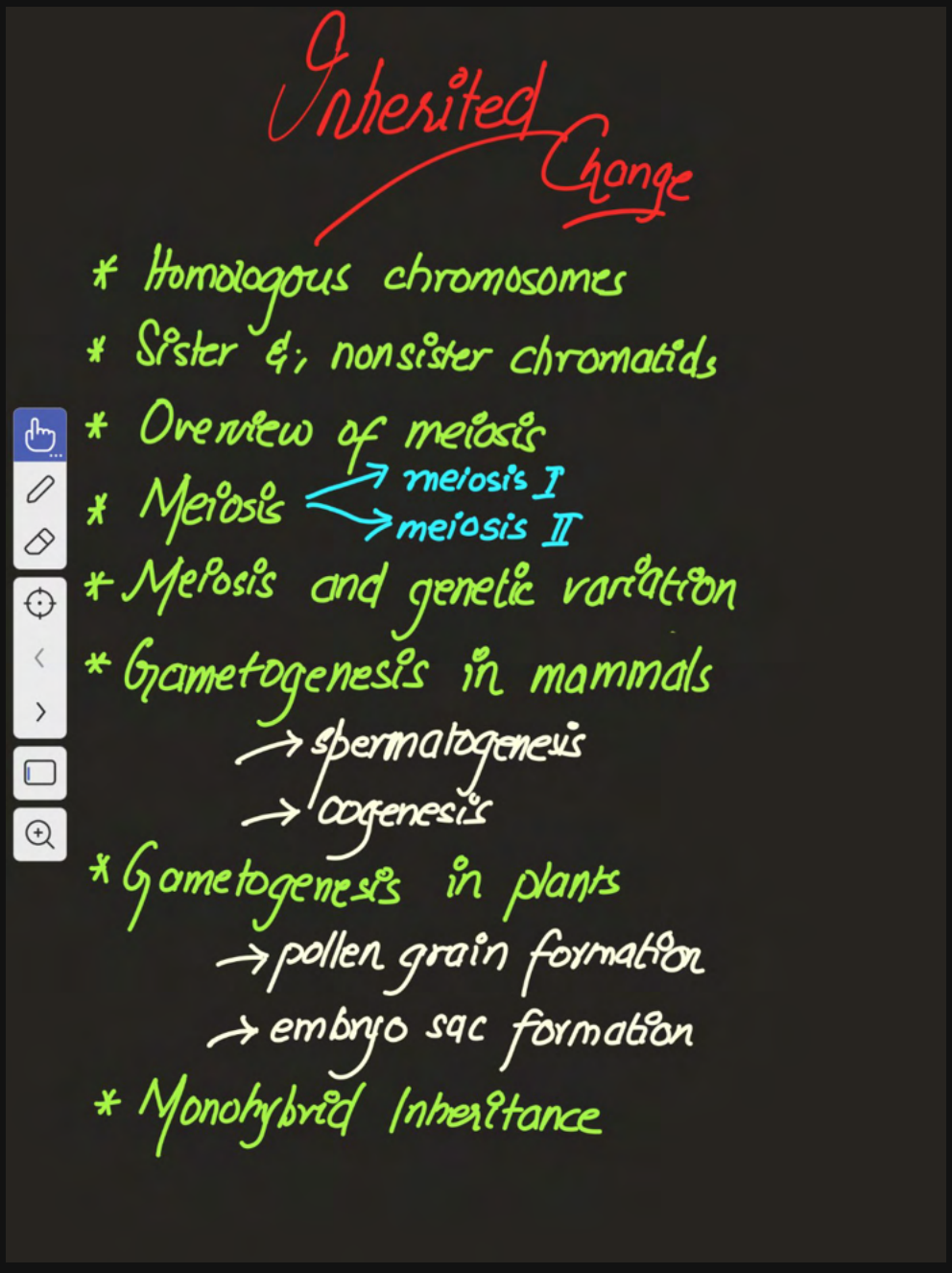
Inherited Change



With

Mohammad Hussham Arshad, MD

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ADVANCED LEVEL BIOLOGY 9700
UNIT 16: Inherited Change

Learning Objectives:

- **Monohybrid Inheritance II**

Video Lecture 6 Slides

Mohammad Hussham Arshad, MD
Biology Department

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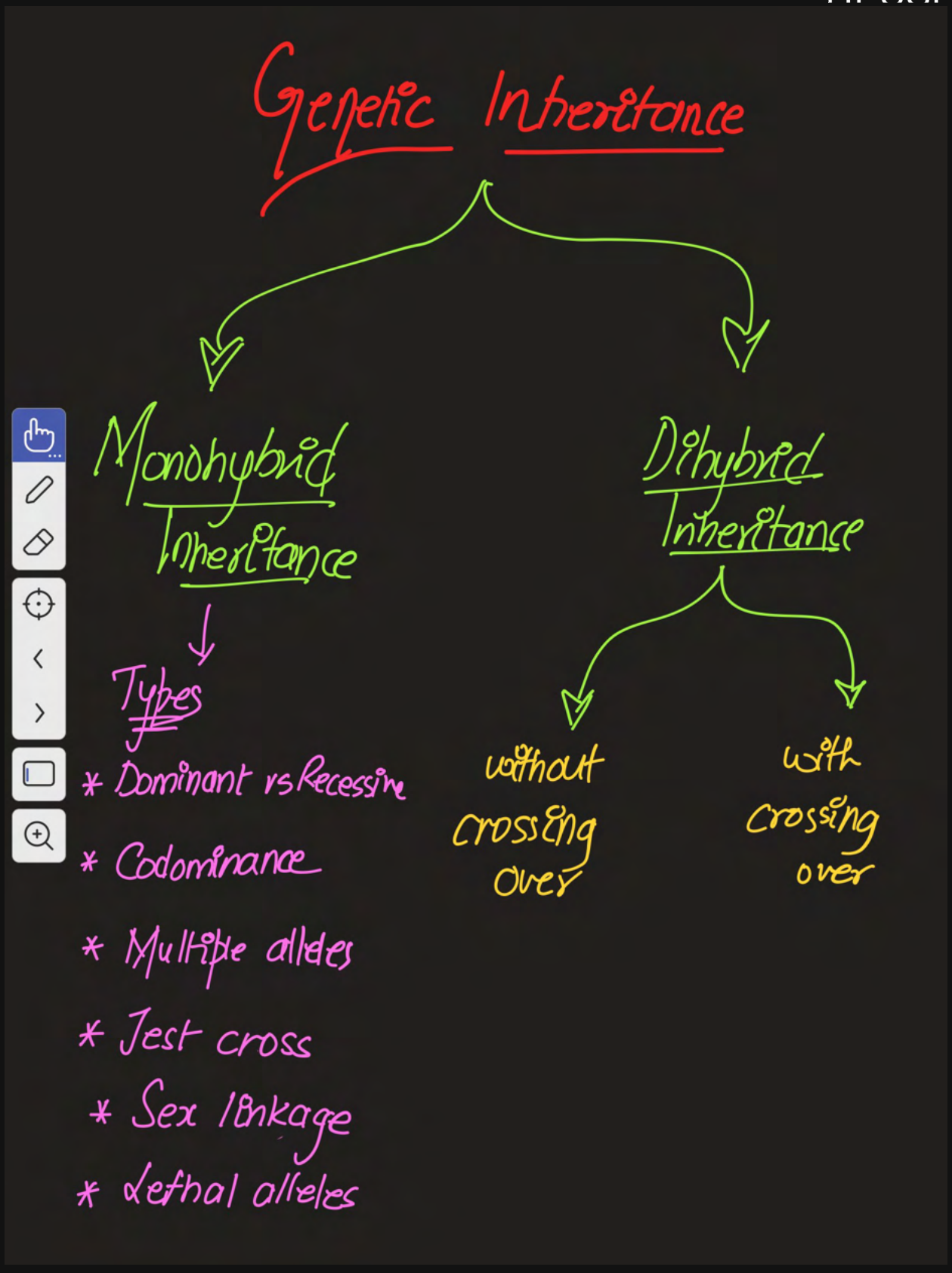
Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis
* Meiosis → meiosis I, meiosis II
* Meiosis and genetic variation
* Gametogenesis in mammals
  - → spermatogenesis
  - → oogenesis
* Gametogenesis in plants
  - → pollen grain formation
  - → embryo sac formation
* Monohybrid Inheritance


Genetic Inheritance
Monohybrid Inheritance
Dihybrid Inheritance
Types
* Dominant vs Recessive
* Codominance
* Multiple alleles
* Test cross
* Sex linkage
* Lethal alleles
without crossing over
with crossing over

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(B) Co-dominance

(1) Two alleles are said to be co-dominant if they express themselves equally in the phenotype. The phenotype of the hetero-zygote is different from homozygotes.

(2) fig. 3A/3B shows the example of snap dragon plants which have 2 equally dominant alleles which determine flower colour, C^R for red colour and C^W for white colour.

AA Aa aa

③ The letter "C" indicates the gene locus for flower colour whereas the superscripts R & W refer to the alleles.

④ A snapdragon plant with red flowers has the genotype C^RC^R. A snapdragon with white flowers has the genotype C^WC^W. A snapdragon plant having genotype C^RC^W has flowers with red & white patches (such that the flowers appear pink in colour)

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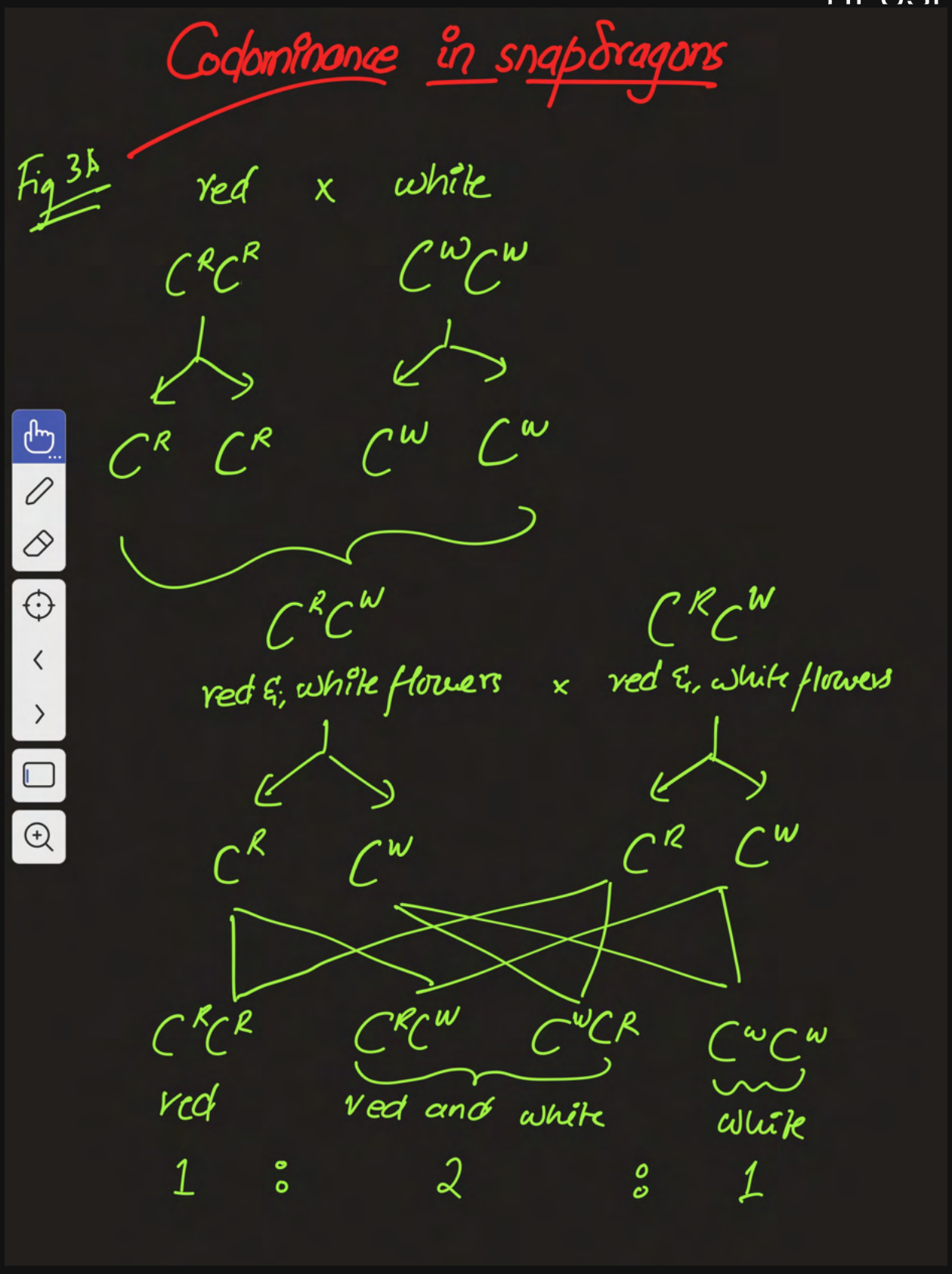
Codominance in snapdragons

Fig 3A



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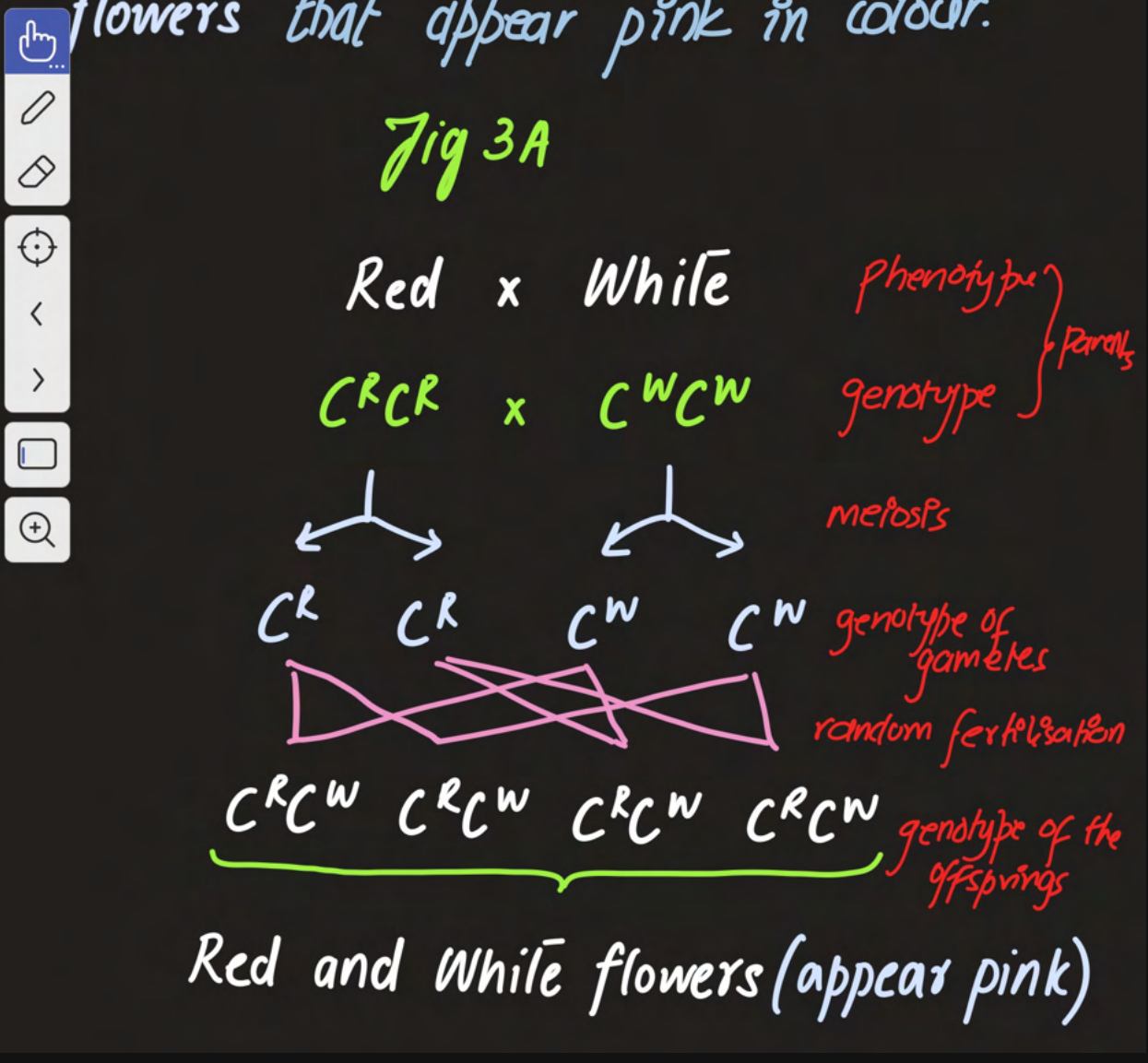
⑤ Fig 3A shows a cross b/w a plant with red flowers and a plant with white flowers. The offspring plants have flowers that appear pink in colour.

Fig 3A



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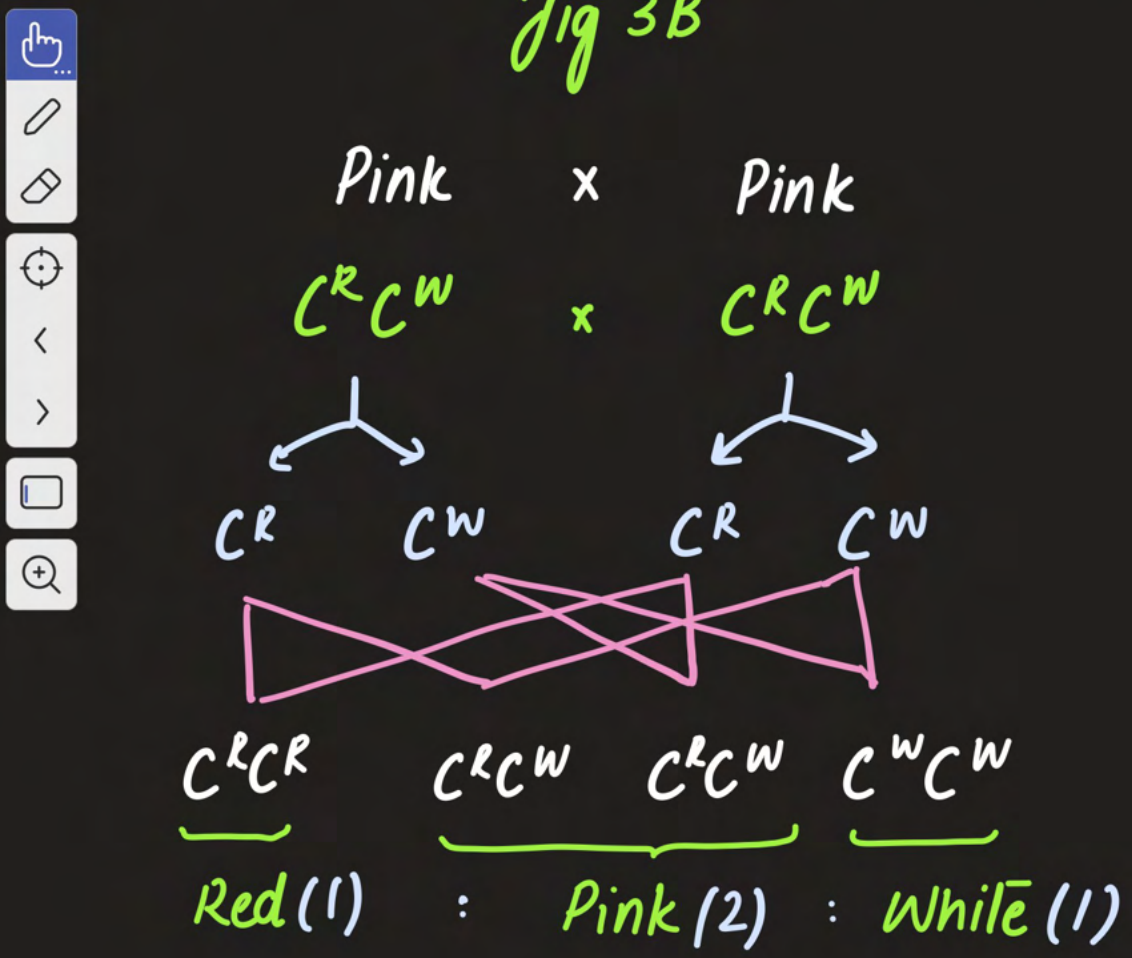
(b) Jig 3B shows a cross b/w 2 heterozygotes The offspring plants have The following phenolype ratio 1 : 2 : 1 (red : pink : white)

Jig 3B



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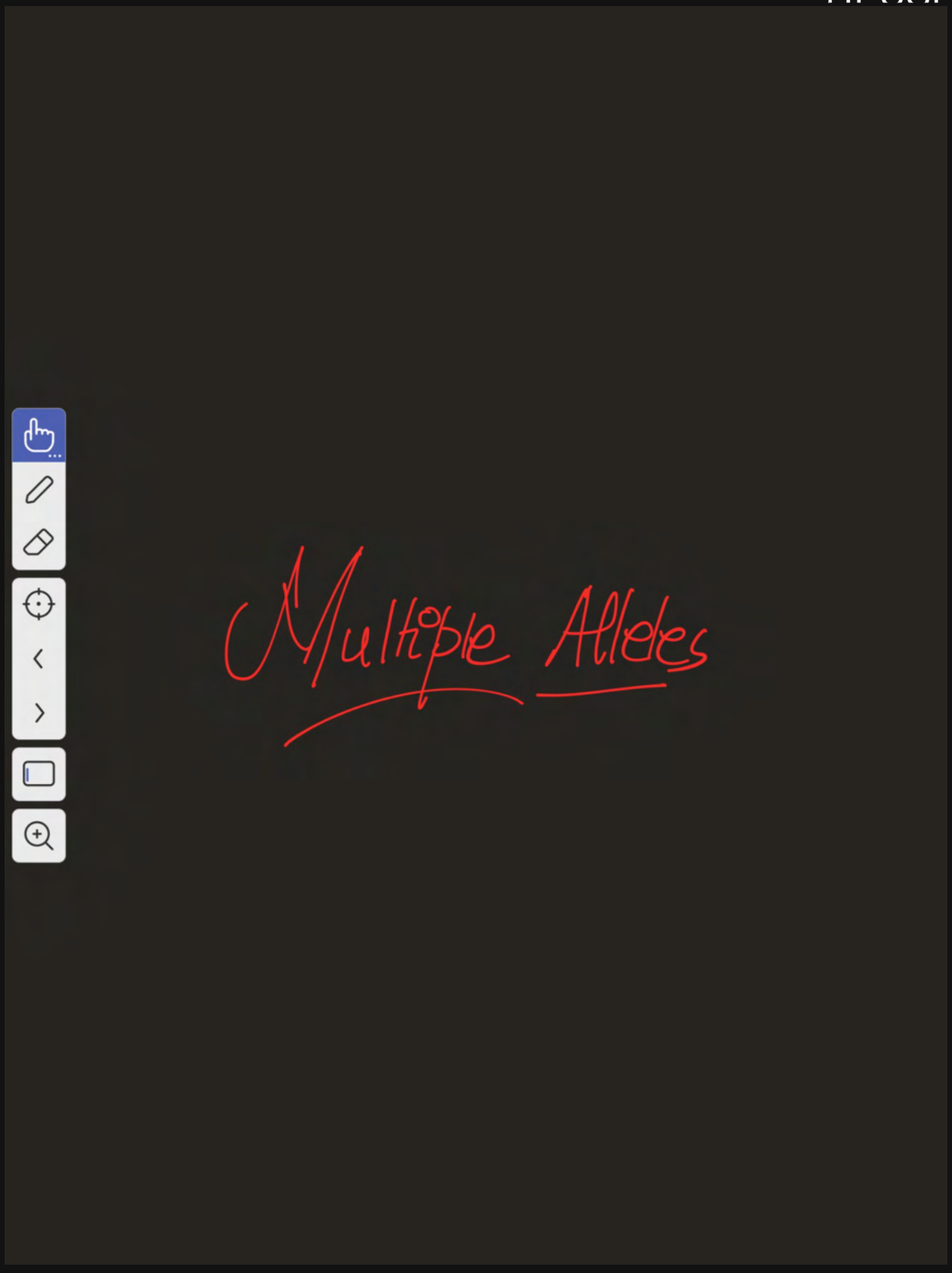

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C Multiple Alleles

1. A gene may have more than 2 alleles. This phenomenon is known as multiple alleles.
2. Alleles that determine the ABO grouping are an example of multiple alleles.
3. The alleles responsible for ABO blood groups are I^A, I^B, I^O where the letter "I" corresponds to the gene locus and the letters A, B and O correspond to the alleles.

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(4) Fig 4 shows a cross b/w 2 heterozygotes having a blood group A (I^AI^O) and a blood group B (I^BI^O).

) The offspring have an equal likelihood of having a blood group A, B, AB or O.

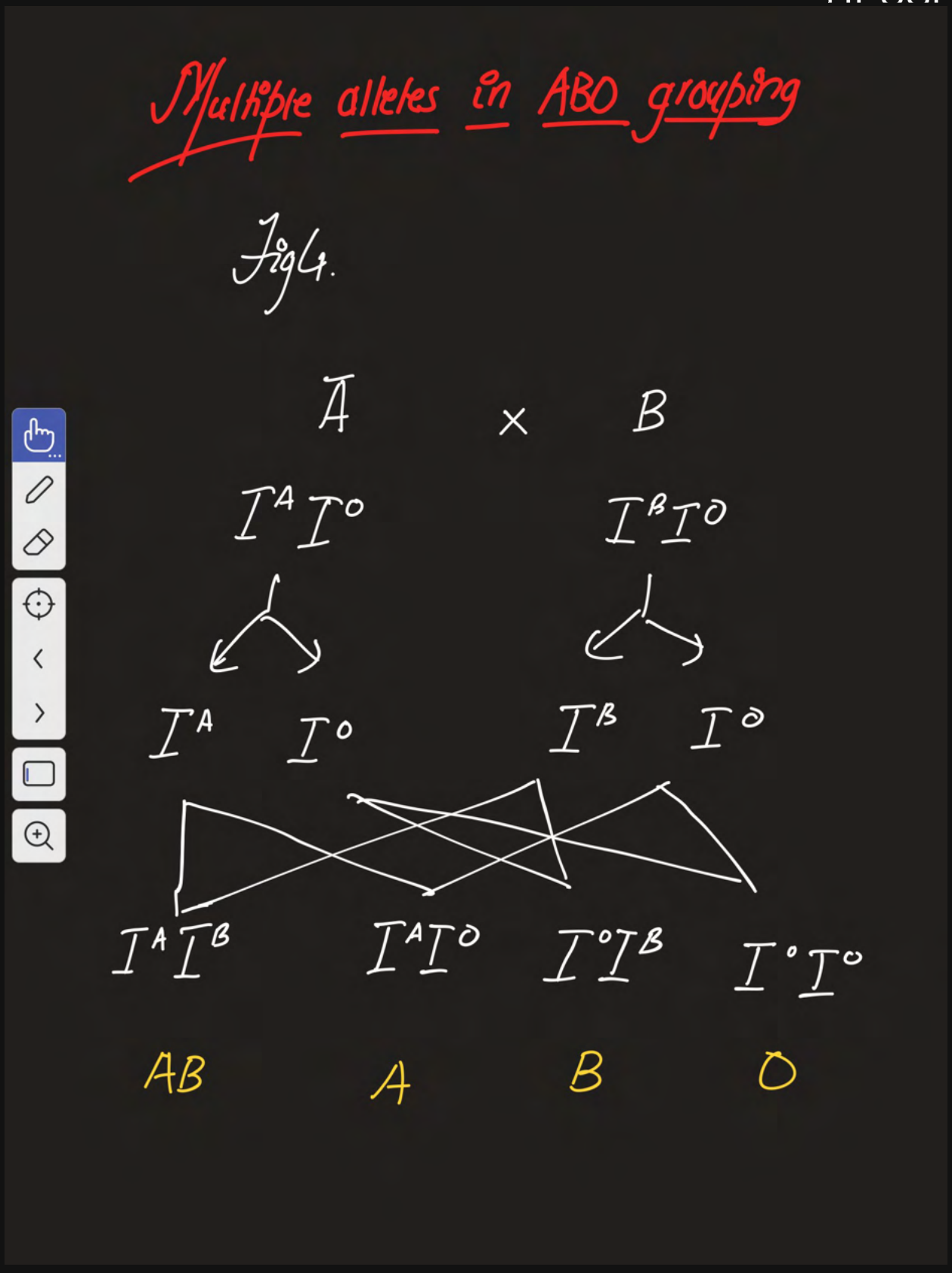
Multiple alleles in ABO grouping
Fig 4.
A
×
B
$I^A I^O$
$I^B I^O$
I^A
I^O
I^B
I^O
$I^A I^B$
$I^A I^O$
$I^O I^B$
$I^O I^O$
AB
A
B
O

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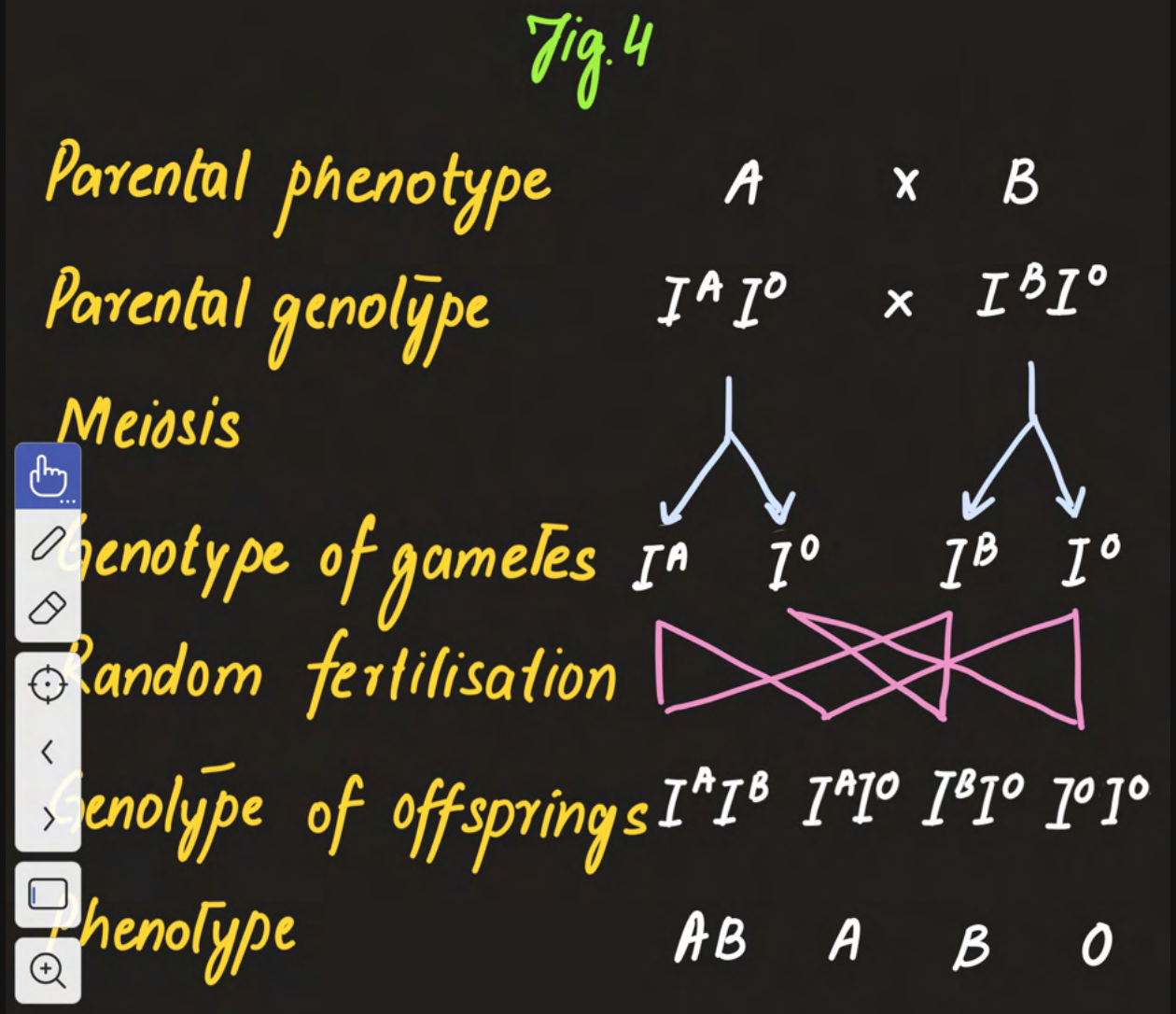
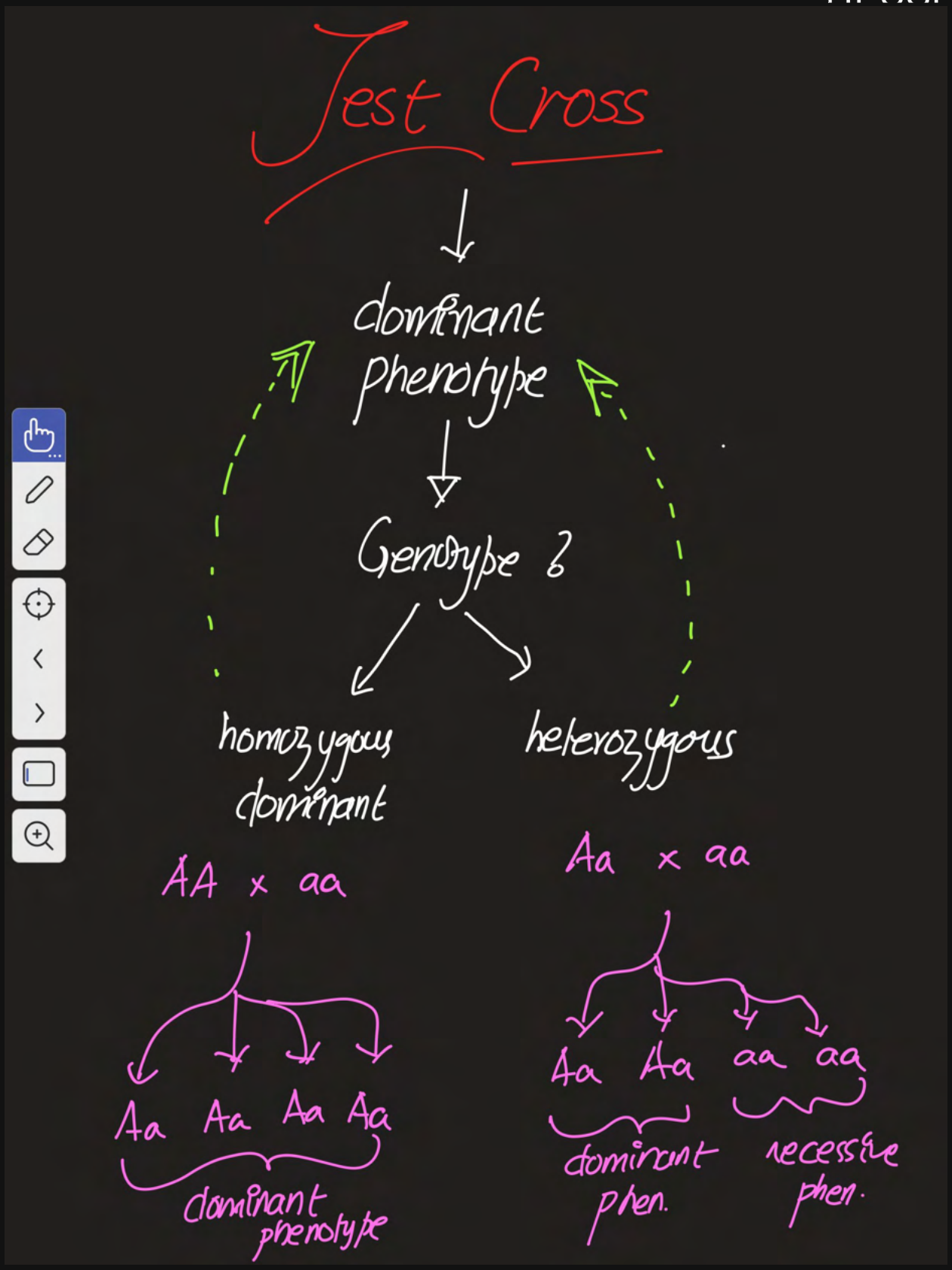
Fig. 4


Parental phenotype
A x B
Parental genotype
$I^A I^O$ x $I^B I^O$
Meiosis
Genotype of gametes
I^A I^O I^B I^O
Random fertilisation
Genotype of offsprings
$I^A I^B$ $I^A I^O$ $I^B I^O$ $I^O I^O$
Phenotype
AB A B O


Test Cross
dominant phenotype
Genotype ?
homozygous dominant
heterozygous
AA x aa
Aa Aa Aa Aa
dominant phenotype
Aa x aa
Aa Aa aa aa
dominant phen.
recessive phen.

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⑤ Test Cross

① For alleles that show dominance, it is not possible to tell the genotype of an organism having dominant phenotype. In such cases, a test cross may be used to determine the unknown genotype.

② A Test cross is a genetic cross b/w an organism with a dominant phenotype and an other organism which is homozygous recessive. The phenotype of the offspring can be used to determine if the genotype

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is homozygous dominant or heterozygous.

③ Consider the genetic diagrams shown in Fig 5a and Fig 5b

④ If a cross b/w a tall pea plant and a short pea plant produces offsprings that are all tall (Fig 5a), it implies that the genotype of the tall organism is homozygous dominant.

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⑤ If a cross b/w a tall pea plant and a short pea plant produces offsprings that are tall & short (Fig 5b), it implies that the genotype of the tall plant is hetero-zygous.

Fig 5a

TT x tt

T T t t

Tt Tt Tt Tt

ALL Tall

Fig 5b

Tt x tt

T t t t

Tt Tt tt tt

tall : short

1 : 1

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Sex-linkage

* a gene that is present on the part of the X-chromosome unmatched by the Y-chromosome is said to be sex-linked.
* A male (in humans) will only have one copy of the gene.
* A female (in humans) will have two copies of the gene

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(E) Sex linkage

(1) Sex linkage refers to the phenomenon of having a gene on the part of the X chromosome not matched by the Y chromosome. Males therefore only have one copy of the gene and females have two copies of the gene.

(2) Males (humans) are heterogametic which implies that they produce gametes having different sex chromosomes.

③ Females (humans) are homogametic which implies that they produce gametes having the same type of sex chromosomes.

④ Haemophilia is a sex linked recessive disorder.

⑤ The haemophilia allele is linked To the X-chromosome. The dominant allele is the normal allele denoted by X^H.

The recessive haemophilia allele is denoted by X^h.

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⑥ The normal allele (X^H) codes for a functional clotting factor VIII. The haemophilia allele (X^h) codes for a non-functioning clotting factor VIII.

⑦ Fig 6a shows genetic cross b/w a haemophiliac male (X^hY) and a carrier female (X^HX^h). The offsprings have a 50% likelihood of having haemophilia.

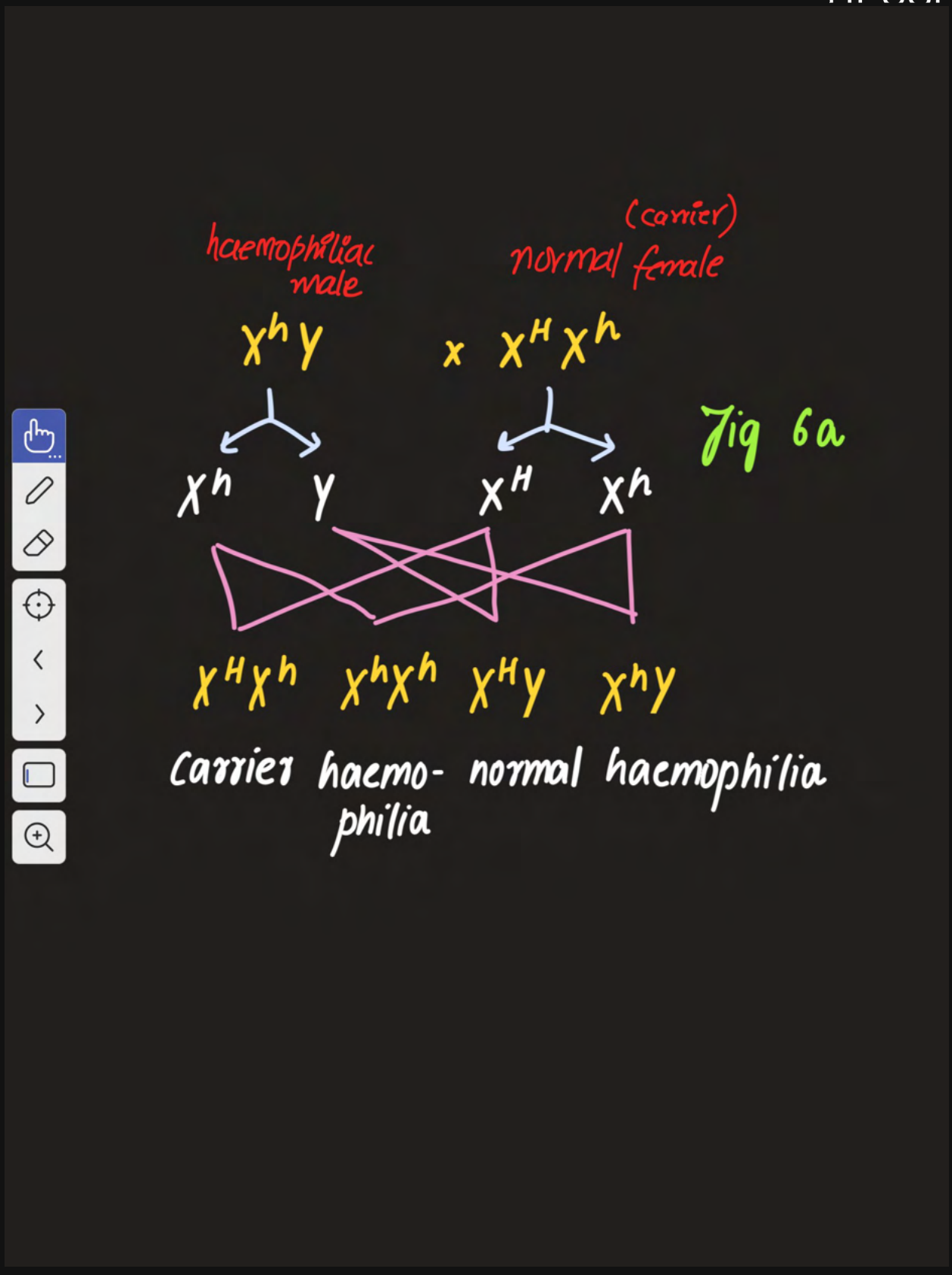
haemophiliac male
normal female (carrier)
X^hY x X^HX^h
X^h Y X^H X^h
X^HX^h X^hX^h X^HY X^hY
Carrier
haemo-philia
normal
haemophilia

Fig 6a

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⑧ Fig 6b shows a genetic cross b/w a carrier female and a normal male. Fig 6a and 6b indicate that a man with haemophilia cannot pass the haemophilia allele to his son but may pass it to his grandson.

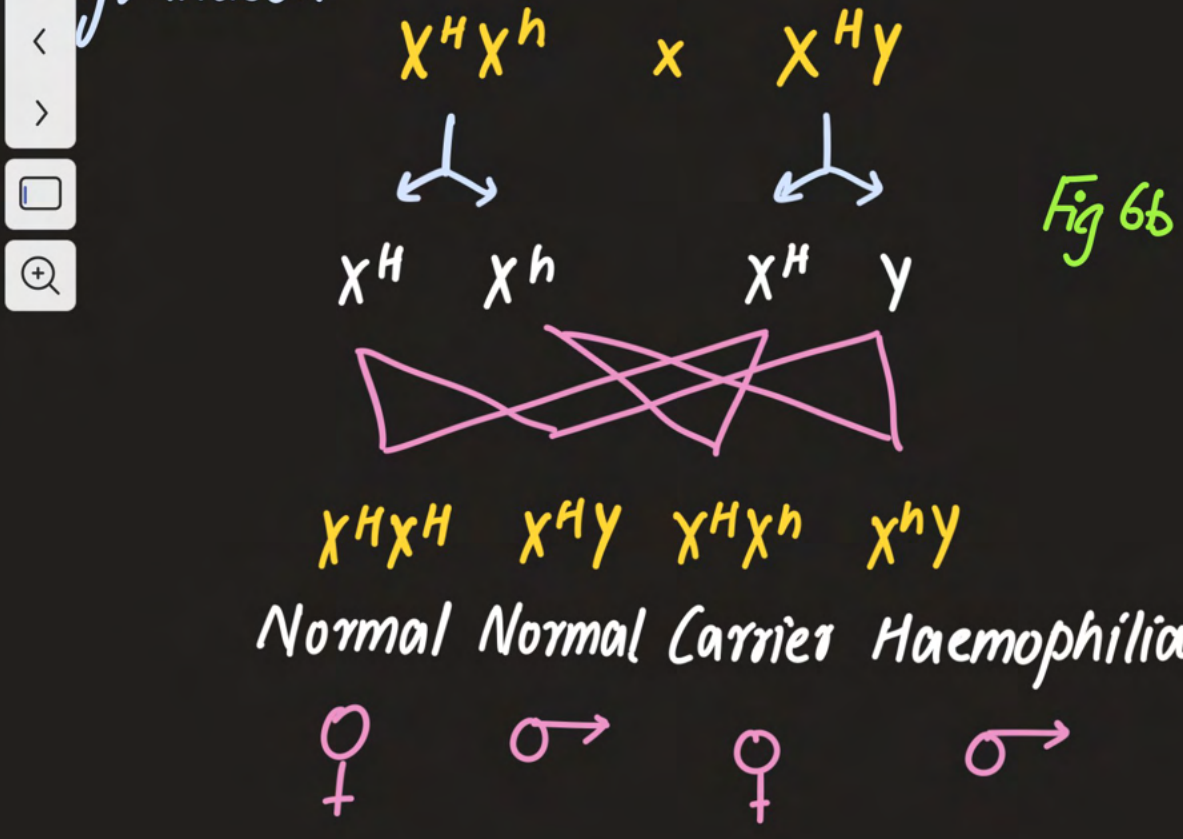

Fig 6b

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Q: Explain with the help of genetic diagram why a male with haemophilia cannot pass haemophilia allele to his son but may pass to his grandson?

Ans: Haemophilia is a sex-linked recessive disorder. The haemophilia allele is present on the X-chromosome. Males being heterogametic (XY) have only one copy of the allele. Females being homogametic (XX) have 2 copies of the allele.

A man with haemophilia has the genotype X^hY. A haemophiliac male can pass on the haemophilia allele to his daughter but not to his son as shown in the figure below (6a):

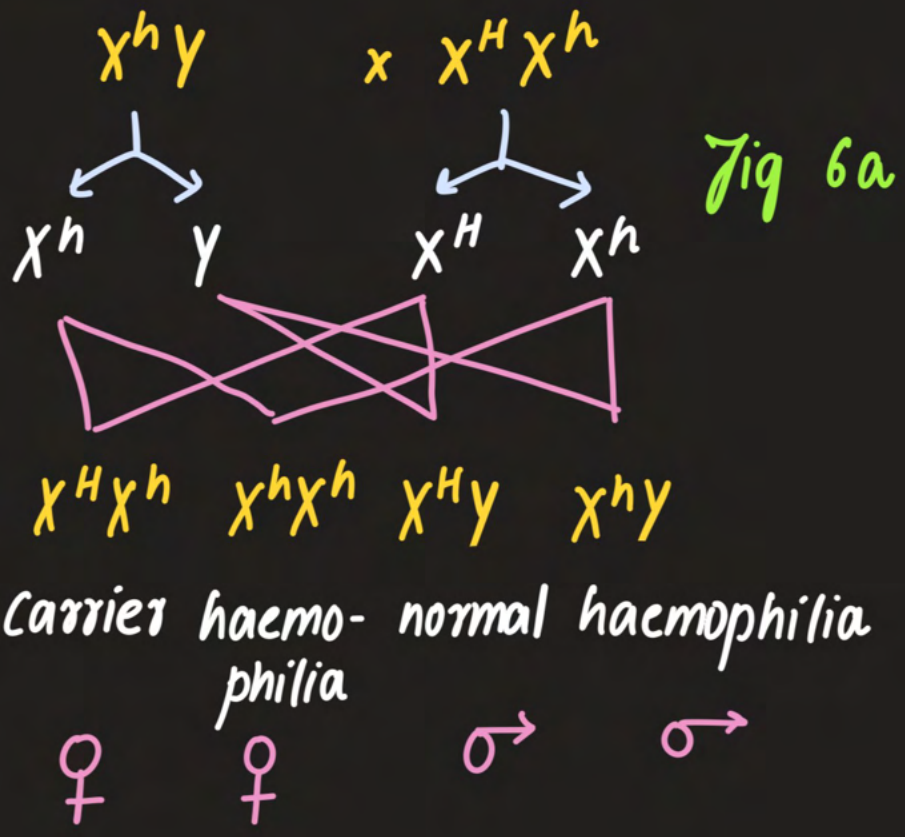

Fig 6a

The carrier female in the cross shown above may pass on the haemophilia allele to her son as shown in the genetic diagram below (6b):

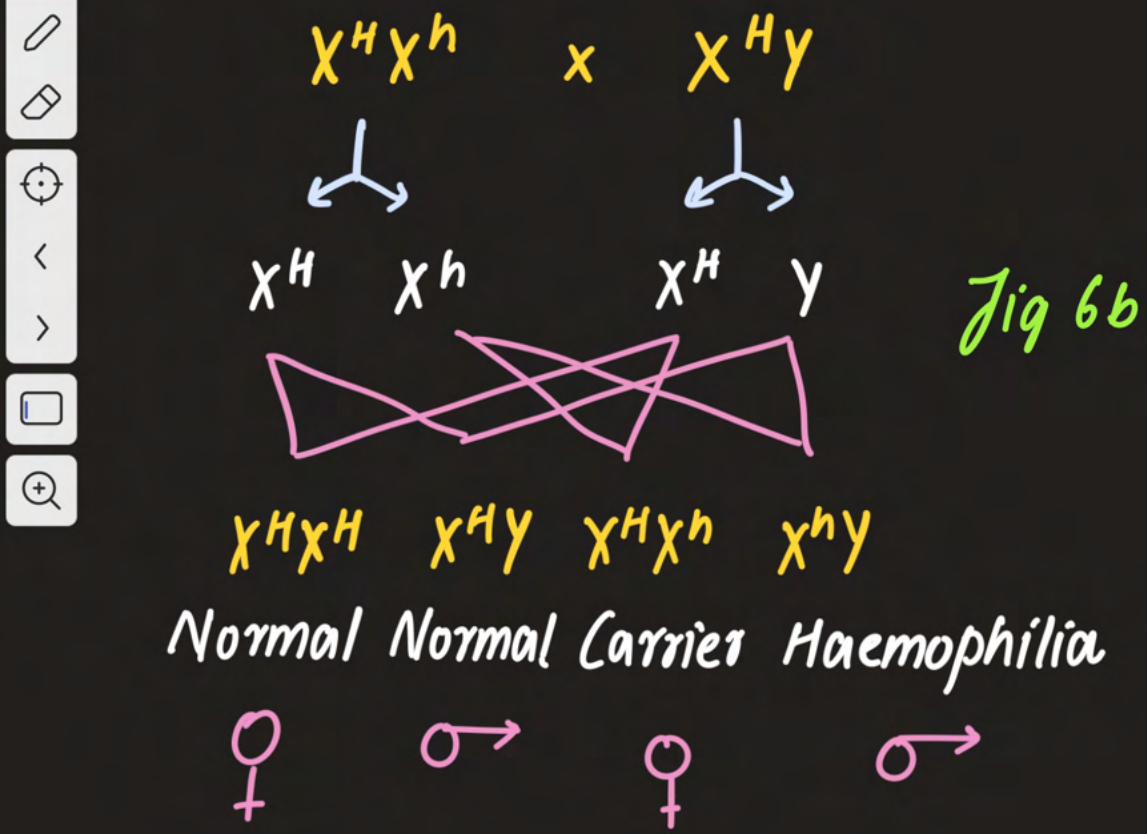

Fig 6b


Questions

Q.

3 Some neurones in the brain produce a neurotransmitter known as dopamine. Parkinson's disease occurs when the neurones that produce dopamine die. A person with the disease may experience difficulty in coordinating movement, often seen as tremors (shaking) in different parts of the body.

Parkinson's disease typically occurs in people older than 55 years. Younger people with these symptoms are said to have early onset Parkinson's disease (EOPD).

Recessive mutations in a gene known as *PINK1*, located on chromosome 1, an autosome, are believed to be one cause of EOPD. A person with this form of EOPD has a homozygous recessive genotype.

(a) Draw a genetic diagram of a cross between two individuals who are heterozygous at the *PINK1* gene locus.

key to symbols used for alleles

E → normal dominant allele

e → recessive EOPD allele

parental genotypes Ee x Ee

gametes E e E e

offspring genotypes EE Ee Ee ee

normal EOPD

ratio of offspring phenotypes 3 : 1

[4]

4 Domestic goats, *Capra hircus*, show a wide range of coat patterns and colours. One gene involved in coat colour and pattern has multiple alleles. Four of these alleles are:

- **A**, the allele for white, is dominant to all others
- $\mathbf{A^b}$, the allele for badgerface (stripes on face) and $\mathbf{A^g}$, the allele for grey, are codominant
- **a**, the allele for black, is recessive to all others.

(a) State **all** the possible genotypes of:

a grey badgerface goat ..... A^bA^g

a white goat. ..... AA, Aa, AA^g, AA^b

[2]

(b) A cross between a black goat and a white goat produced a white goat. This white offspring was crossed with a grey goat. The genotype of the grey goat was not known.

Use genetic diagrams to show **all** the possible offspring genotypes and phenotypes that could result from the cross between the white offspring and the grey goat.

white goat (A_) × black goat (aa) → white offspring (Aa)

White offspring × grey goat

cross 1: Aa × A^gA^g

offspring genotypes → AA^g and A^ga

" " phenotypes → white and grey

cross 2: Aa × A^ga

offspring genotypes → AA^g, Aa, A^ga, aa

" phenotypes → white, grey, black

[4]

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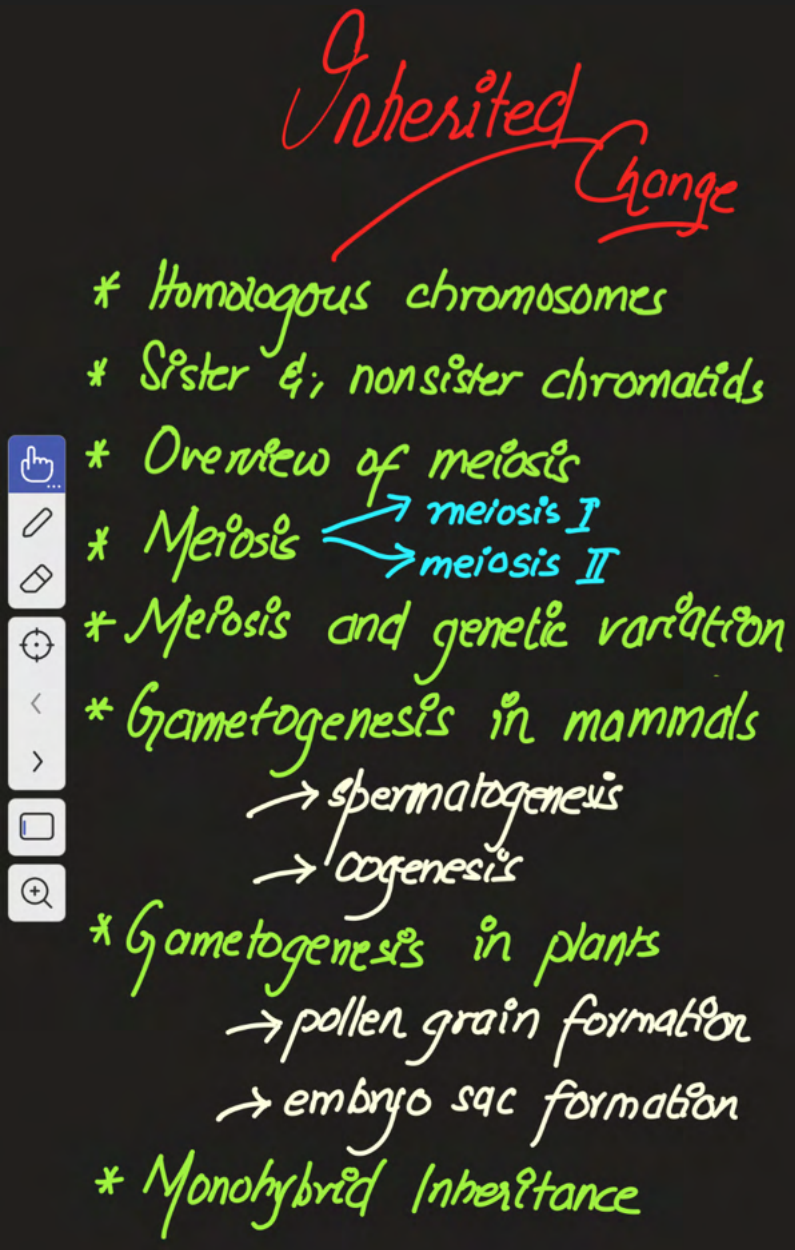
Inherited Change



With

Mohammad Hussham Arshad, MD

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ADVANCED LEVEL BIOLOGY 9700

UNIT 16: Inherited Change

Learning Objectives:

- **Monohybrid Inheritance III**
- **Dihybrid Inheritance I**

Video Lecture 7 Slides

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Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis
* Meiosis → meiosis I, meiosis II
* Meiosis and genetic variation
* Gametogenesis in mammals
  - → spermatogenesis
  - → oogenesis
* Gametogenesis in plants
  - → pollen grain formation
  - → embryo sac formation
* Monohybrid Inheritance

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Genetic Inheritance

Monohybrid Inheritance

Types

* Dominant vs Recessive
* Codominance
* Multiple alleles
* Test cross
* Sex linkage
* Lethal alleles

Dihybrid Inheritance

- without crossing over
- with crossing over

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Questions

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Q.

8 An autosomal gene controlling fur colour in cats has three alleles:

- allele C^B codes for black fur
- allele C^{CH} codes for chocolate fur
- allele C^{CM} codes for cinnamon (orange-brown) fur.

Allele C^B is dominant to both C^{CH} and C^{CM}.
Allele C^{CH} is dominant to C^{CM}.

(a) (i) State the name given to the position of a gene on a chromosome.

locus [1]

(ii) A cinnamon-coloured cat must be homozygous.

Explain what is meant by the term *homozygous*.

* identical alleles of a gene

$C^BC^{CH} \times C^BC^{CM}$ [1]

(b) Two black cats were crossed. Most of their offspring had black fur and a few had chocolate fur. When they were mature, one of the black offspring was crossed with one of the chocolate offspring. Some of the cats in the second generation had cinnamon fur.

Draw a genetic diagram to show how cats with cinnamon-coloured fur were produced in the second generation.

$C^BC^{CH} \times C^BC^{CM} \rightarrow C^BC^B / C^BC^{CM} / C^BC^{CH} / C^{CH}C^{CM}$

parental phenotype: black cat × chocolate cat

parental genotype: C^BC^{CM} × $C^{CH}C^{CM}$

meiosis

gametes: C^B C^{CM} | C^{CH} C^{CM}

offspring genotype: C^BC^{CH} C^BC^{CM} $C^{CH}C^{CM}$ $C^{CM}C^{CM}$

offspring phenotype: black black chocolate cinnamon

[6]

[Total: 8]

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Q.

1 **(a)** Diabetes insipidus (DI) is a condition that results in excessive thirst and the excretion of large amounts of dilute urine. A main cause of DI is a deficiency of ADH.

ADH normally binds to receptor proteins on cell surface membranes of cells in the collecting ducts of nephrons.

Outline the effect of ADH on the collecting ducts.

.....

.....

.....

.....

.....

.....

.....

..... [3]

(b) An inherited form of DI may be caused by faulty membrane receptors. These receptors are coded for by a sex-linked allele.

(i) Explain the term *sex linkage*.

* allele is present on the X-chromosome

.....

.....

..... [1]

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(ii) Use a genetic diagram to show how parents who do not have this condition can have a child with the inherited form of DI.

symbols

normal → X^D

recessive DI → X^d

parental genotypes $X^DY \times X^DX^d$

gametes X^D Y X^D X^d

offspring genotypes X^DX^D X^DX^d X^DY X^dY

offspring phenotypes normal female | normal female (carrier) | normal male | DI male

[4]

[Total: 8]

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Lethal Alleles

↓

alleles that cause death of an organism are termed as lethal alleles

dominant lethals

↓

if the allele causes DEATH in homozygous state or the heterozygous state

recessive lethals

↓

if the allele causes DEATH only in the homozygous state

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(F) Lethal Alleles

(1) Lethal alleles are alleles that cause the death of an organism.

(2) lethal alleles are classified into recessive lethals and dominant lethals.

(3) Recessive lethals are lethal alleles that cause the death of an organism in the homozygous state. Dominant lethals are lethal alleles that cause the death of an organism in the homozygous or heterozygous state.

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④ lethal alleles were first discovered while studying the inheritance of coat colour in a certain breed of mice.

⑤ The gene for coat colour in this breed of mice has two alleles:

- Y → yellow coat colour
- y → grey coat colour

$Yy \times Yy$

$YY \times YY$

$YY \times Yy$

⑥ Fig 7 shows a cross blw 2 yellow mice which always produces offsprings in the ratio 2 : 1 for yellow : grey mice. This ratio does not obey the expected phenotypic

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ratio.

⑦ The observed phenotypic ratio results because the dominant allele Y is a lethal allele which causes death of an organism which is homozygous dominant, therefore yellow mice can only have the genotype Yy.

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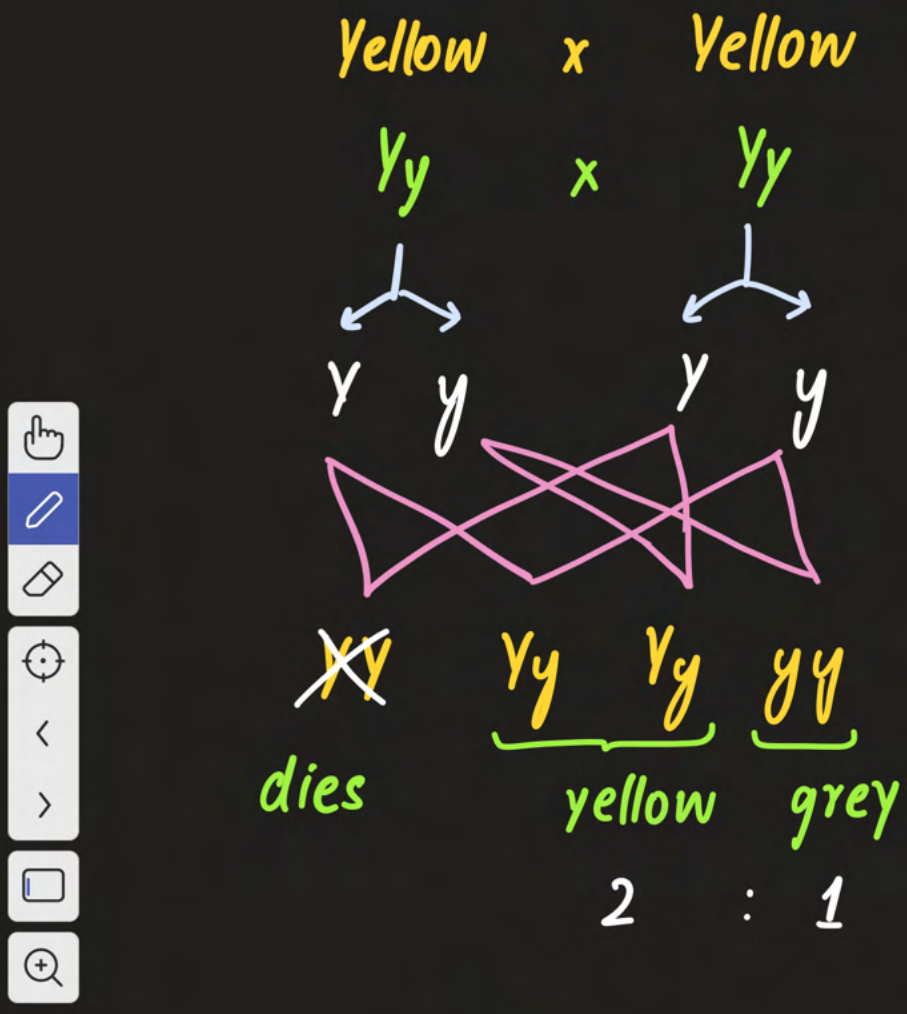

Fig 7

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Q.

6 In mice, fur colour is controlled by a gene with multiple alleles. These alleles are listed in no particular order.

black and tan = C^{bt}	yellow = C^{y}
agouti = C^{a}	black = C^{b}

(a) Suggest explanations for the results of the following crosses between mice.

(i) Mice with agouti fur crossed with mice with black fur may produce all agouti offspring **or** some agouti and some black offspring.

Agouti x Black → all agouti
→ agouti + black

$C^aC^a \times C^bC^b$ / $C^aC^b \times C^bC^b$

* agouti allele is dominant to black allele
* agouti parent may be homozygous or heterozygous

[2]

(ii) Crosses between heterozygous parents with the genotype $C^{y}\ C^{b}$ always produce a ratio of two yellow mice to one black mouse.

$C^yC^b \times C^yC^b$

$C^y\ C^b$ $C^y\ C^b$

C^yC^y C^yC^b C^yC^b C^bC^b

C^yC^y dies in utero; C^yC^b C^yC^b : C^bC^b = 2 : 1

* yellow allele is dominant to black allele
* the homozygous genotype C^yC^y is lethal [2]

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(iii) Mice with yellow fur crossed with mice with black fur will produce one following outcomes:

- some yellow offspring and some agouti offspring
- some yellow offspring and some black and tan offspring
- some yellow offspring and some black offspring.

yellow × black

C^yC^b C^bC^b

* yellow allele C^y is dominant to all others
* all yellow mice are heterozygous

[2]

(b) A test cross is used to determine the genotype of an organism.

Describe how you would carry out a test cross to determine the genotype of a black and tan mouse.

* black and tan mouse will be crossed with a homozygous recessive black mouse C^bC^b
* If all offsprings are black and tan → $C^{bt}C^{bt}$
* If some offsprings are black and some are black and tan → $C^{bt}C^b$

[2]

[Total: 8]

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Bihybrid Inheritance

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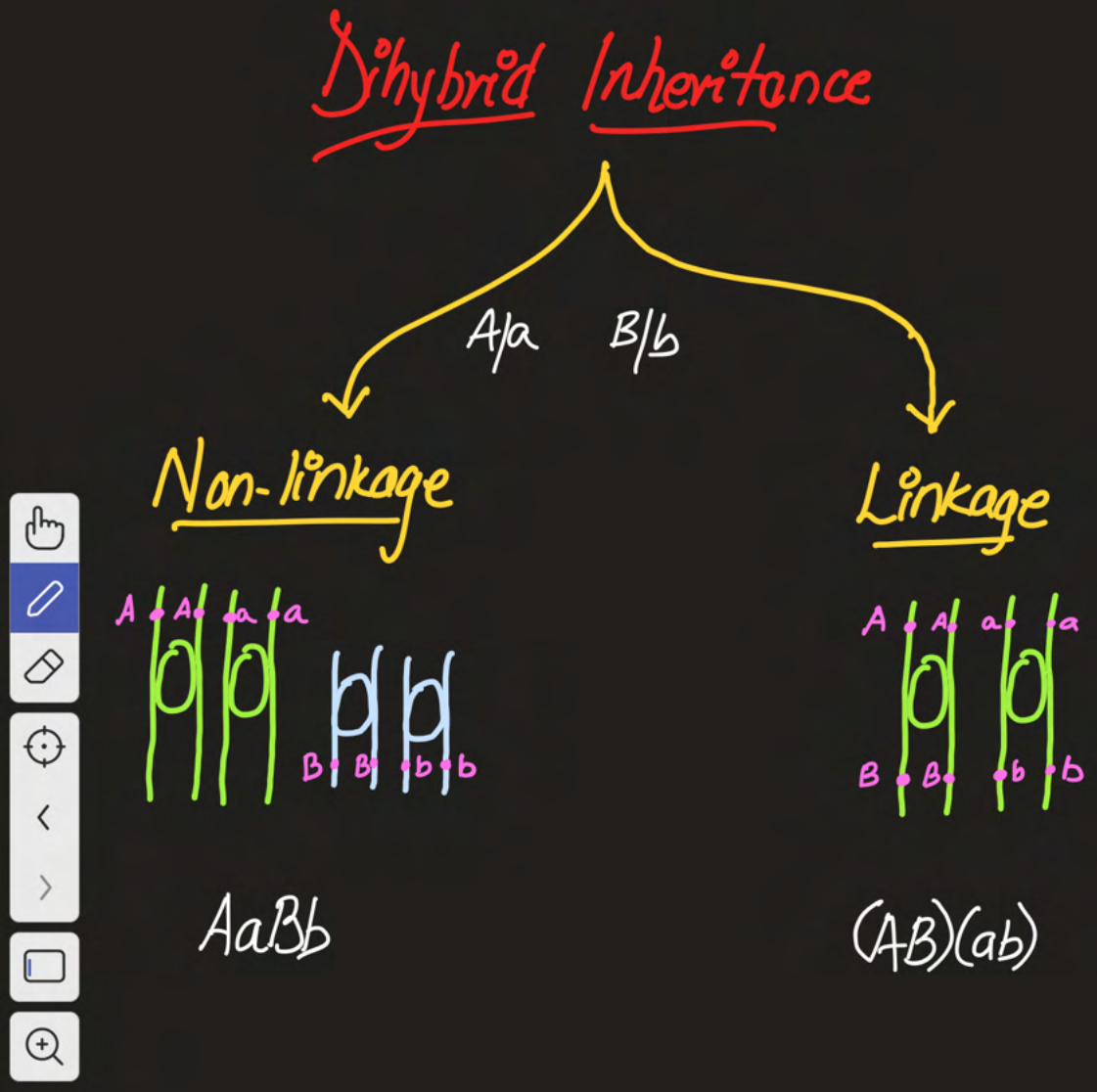
Dihybrid Inheritance
A/a B/b
Non-linkage
Linkage
A A a a
B B b b
AaBb
A A a a
B B b b
(AB)(ab)

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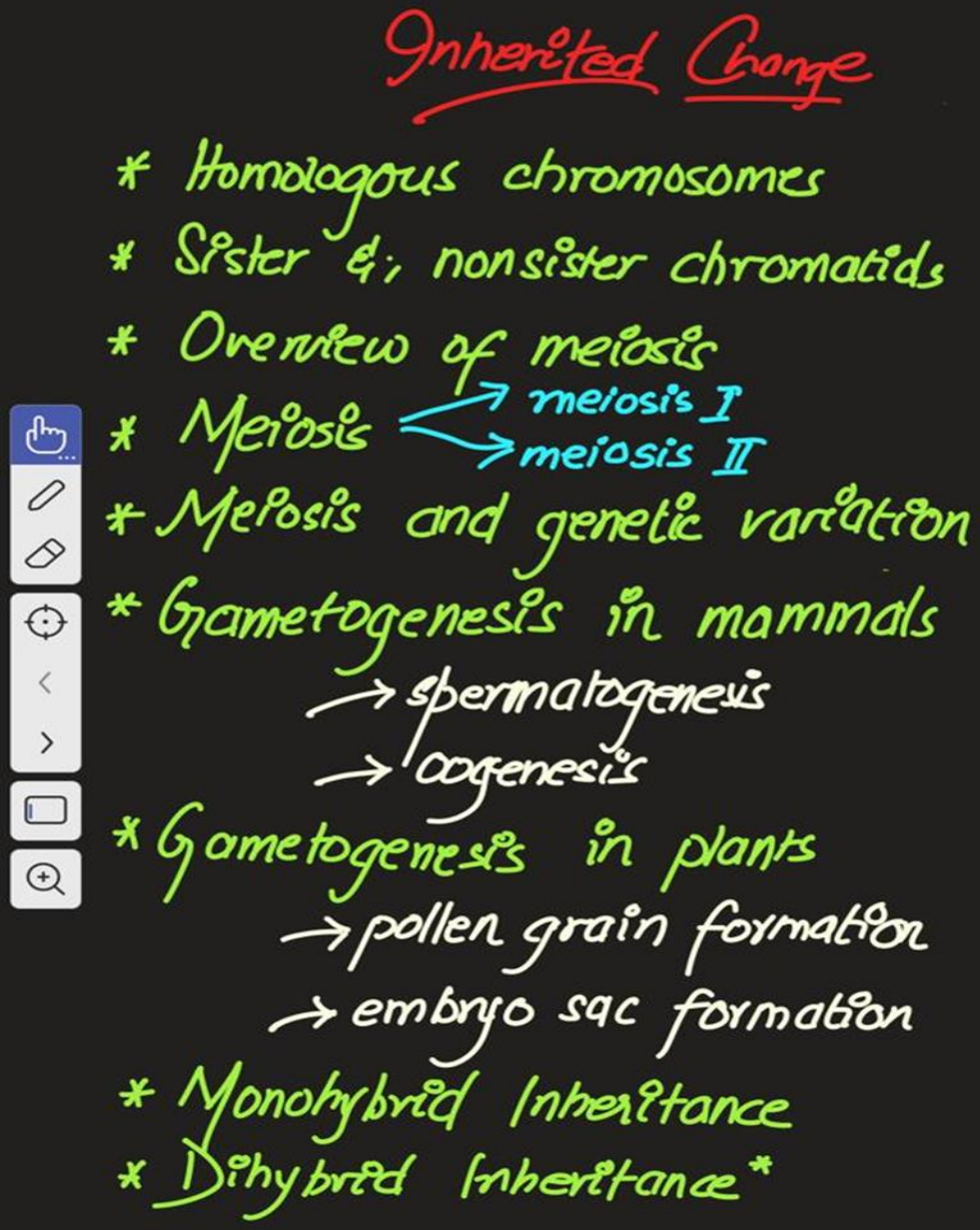
INHERITED CHANGE



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Inherited Change

* Homologous chromosomes
* Sister & nonsister chromatids
* Overview of meiosis
* Meiosis → meiosis I, meiosis II
* Meiosis and genetic variation
* Gametogenesis in mammals
  - → spermatogenesis
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* Gametogenesis in plants
  - → pollen grain formation
  - → embryo sac formation
* Monohybrid Inheritance
* Dihybrid Inheritance*

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Dihybrid Inheritance

A/a B/b

Non-linkage

AaBb

* Chromosomes can assort independently

Genotype of gametes:

AB ab aB Ab

1 : 1 : 1 : 1

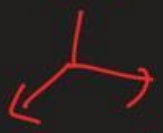
Linkage

(without crossing over)

(AB)(ab)

* Independent assortment does NOT occur

(AB) (ab)



(AB) (ab)

1 : 1

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Dihybrid Inheritance

* refers To inheritance of two genes simultaneously.

(A) Dihybrid Inheritance of Non-linked Genes

- Non- linkage is the phenomenon of having two genes on separate pairs of homologous chromosomes.
- Consider two genes A/a and B/b;

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Q: How do we write the genotype of the organism above?

AaBb

Q: State the genotype and probability of the gametes produced. Explain how they are produced.

Gametes → AB : Ab : aB : ab

1 : 1 : 1 : 1

Gametes are produced as a result of independent assortment.

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(B) Dihybrid Inheritance of Linked Genes without crossing over

- Linkage is the phenomenon of having two genes on the same pair of homologous chromosome. Linked genes CANNOT assort independently.
- Consider Two genes A/a & B/b;

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Q : How do we write the genotype of the organism above?

(AB)(ab)

Q : State the genotype and probability of the gametes produced without crossing over?

Gametes → AB : ab

1 : 1

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© Dihybrid Inheritance of Linked Genes with crossing over

- Crossing over refers to exchange of genetic material b/w non-sister chromatids
- Consider two genes A/a and B/b;

(AB)(ab)

meiosis with no crossing over
- AB
- ab

meiosis with crossing over
- (more common) AB, ab } parental gametes
- (less common) Ab, aB } gametes due to crossing over

'recombinant gametes'

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Question :

- Consider two genes A/a & B/b which determine the seed colour and seed shape respectively:

A - yellow B - smooth

a - green b - wrinkled

Case A → Non-linkage

Case B → Linkage without crossing over

Case C → Linkage with crossing over

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Case A : A/a and B/b are non-linked genes.

- Assume the two organisms are heterozygous for both the genes.

Genotype of parent: AaBb x AaBb

Genotype in gametes (same in both parents) : AB, aB, ab, Ab

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- A Punnett square can be made to determine the genotype and phenotypic ratio of offsprings:

♀ \ ♂	AB 1/4	Ab 1/4	aB 1/4	ab 1/4
AB 1/4	AABB 1/16	AABb	AaBB	AaBb
Ab 1/4	AABb	AAbb	AaBb	Aabb
aB 1/4	AaBB	AaBb	aaBB	aaBb
ab 1/4	AaBb	Aabb	aaBb	aabb

Phenotypes: Yellow & Smooth : Yellow & Wrinkled : Green & Smooth : Green & Wrinkled

Phenotypic ratio: 9 : 3 : 3 : 1

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- We will now discuss for the same two genes (A/a and B/b), the expected outcome of offsprings when:

Case B : The two genes are linked (with NO crossing over)

Case C : The two genes are linked (with crossing over)

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* Before we start, let's look at the two forms of linkage with respect to dihybrid inheritance:

① Autosomal Linkage → The two genes are present on the same chromosome which is NOT a sex chromosome.

② Sex linkage → The two genes are on the X chromosome.

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Case B : A/a and B/b have autosomal linkage with no crossing over

* Again assume the two organisms are heterozygous for both the genes (A/a and B/b)

- Genotype of parent : (AB)(ab) x (AB)(ab)
- Genotype of gametes: AB, ab

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Phenotype of parent: Yellow & Smooth × Yellow & Smooth

Genotype of parent: (AB)(ab) × (AB)(ab)

Meiosis

Genotype of gametes: AB ab AB ab

Random Fertilisation

Offspring Genotype: (AB)(AB) (AB)(ab) (AB)(ab) (ab)(ab)

Phenotype: Yellow & Smooth | Green & Wrinkled

Ratio: 3 : 1

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Case C: A/a and B/b have autosomal linkage with crossing over

* Again assume the two organisms are heterozygous for both the genes (A/a and B/b)
  - Genotype of parent: (AB)(ab) x (AB)(ab)
  - Genotype of gametes: → due to crossing over: Ab, aB (less common) = recombinant gametes
    → not due to crossing over: AB, ab (more common) = non-recombinant gametes

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* The offsprings that result due to recombinant gametes are termed as recombinants. Their phenotype is said to be the recombinant phenotype.

* Recombinant gametes are produced in much lesser numbers than non-recombinant gametes (parental gametes)

* Percentage of recombinants produced can be calculated using the expression:

$$\frac{\text{recombinant}}{\text{total offspring}} \times 100 = \text{cross-over value (COV)}$$

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Questions

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Q1.

3 Flowers of the common morning-glory plant, *Ipomoea purpurea*, can have several different phenotypes. An example of these flowers is shown in Fig. 3.1.

Fig. 3.1

Flower colour in *I. purpurea* is controlled by **two** genes on **different** chromosomes.

Gene **R**/**r**, which codes for a protein involved in pigment production, has 2 alleles:

- the dominant allele, **R**, allows pigment production
- the recessive allele, **r**, prevents pigment production of any colour, resulting in white flowers.

Gene **T**/**t**, which determines the type of pigment produced, has two alleles:

epistasis

- the dominant allele, **T**, results in purple flowers
- the recessive allele, **t**, results in red flowers.

(a) **(i)** Define the term *allele*.

* alternative forms of a gene
* which have a different nucleotide sequence

..... [2]

(ii) Suggest ways in which the expression of allele **R** allows pigment production.

..... [3]

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(b) Complete Fig. 3.2 to show the results of a cross between two *I. purpurea* plants that are **heterozygous at both loci**.

parental genotypes: RrTt × RrTt

parental gametes: RT Rt rT rt × RT Rt rT rt

results of cross:
- show parental gametes in first column and top row
- show offspring genotypes and phenotypes in square boxes

parental gametes → / ↓	RT	Rt	rT	rt
RT	RRTT purple	RRTt purple	RrTT purple	RrTt purple
Rt	RRTt purple	RRtt red	RrTt purple	Rrtt red
rT	RrTT purple	RrTt purple	rrTT white	rrTt white
rt	RrTt purple	Rrtt red	rrTt white	rrtt white

phenotypic ratios: 9 : 3 : 4

purple : red : white

Fig. 3.2

[5]

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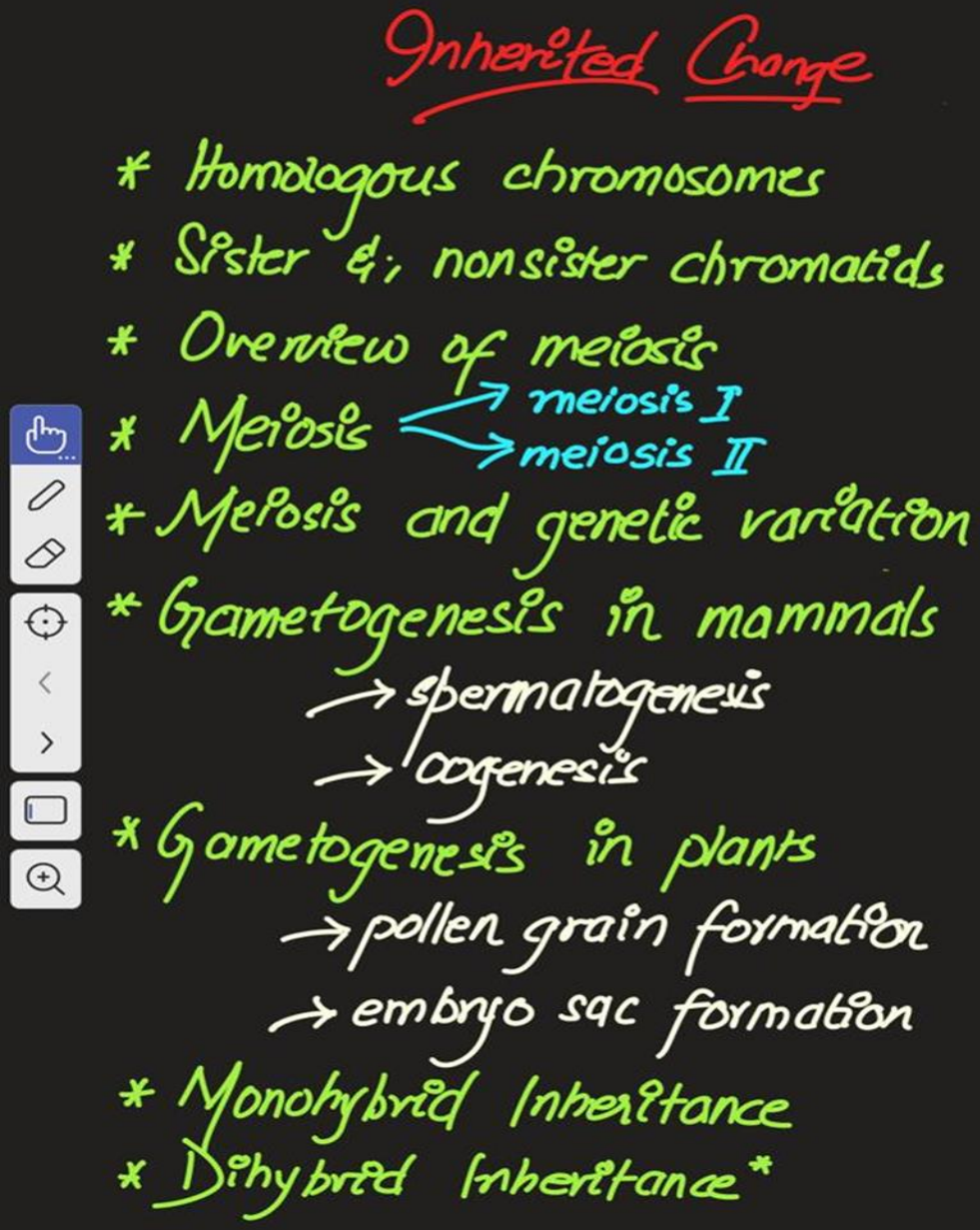

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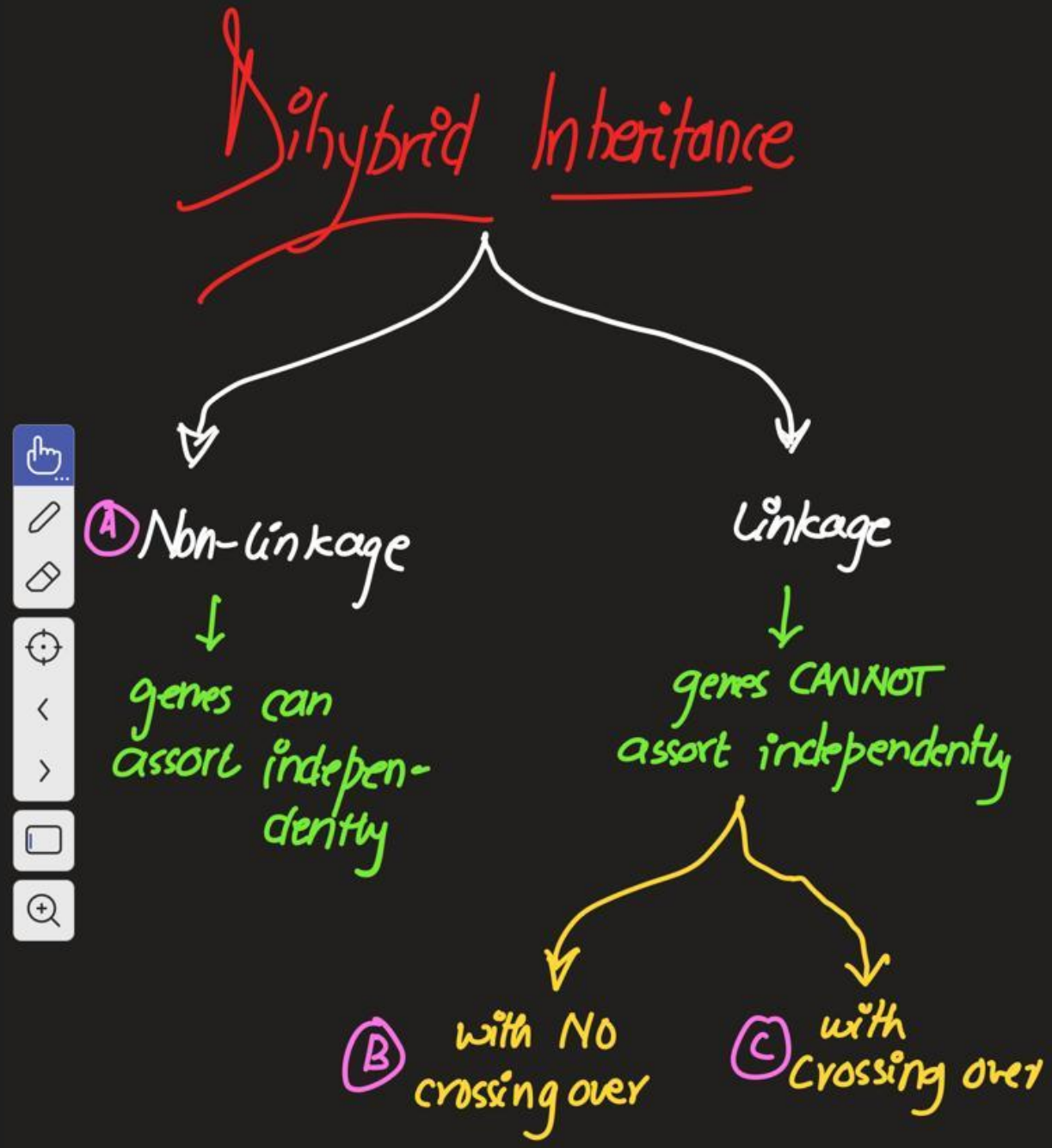
Dihybrid Inheritance
A Non-linkage
genes can assort indepen-dently
Linkage
genes CANNOT assort independently
B with No crossing over
C with Crossing over

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Q2.

2 The patty pan squash plant, *Cucurbita pepo*, produces edible fruits that vary in colour.

(a) The colour of the fruits is controlled by two genes, **A/a** and **B/b**, that occur on different chromosomes.

- Allele **A** produces a white fruit colour.
- Allele **a** does not produce a colour by itself but allows the colours coded by gene **B/b** to show in the phenotype.
- Allele **B** produces a yellow fruit colour.
- Allele **b** produces a green fruit colour.

In a dihybrid cross, an **AABB** plant was crossed with an **aabb** plant. All the resulting F1 plants produced white fruits. AaBb

The F1 plants were then crossed with each other to obtain the F2 generation.

(i) Complete Fig. 2.1 with the gametes produced by the F1 parents, the F2 genotypes and the F2 phenotypes.

State the ratio of fruit colours in the F2 offspring.

F1 parents: **AaBb** × **AaBb**

	AB	Ab	aB	ab
AB	AABB white(w)	AABb w	AaBB w	AaBb w
Ab	AABb w	AAbb w	AaBb w	Aabb w
aB	AaBB w	AaBb w	aaBB yellow	aaBb yellow
ab	AaBb w	Aabb w	aaBb yellow	aabb green

Fig. 2.1

ratio of fruit colours in F2 offspring ..... 12 : 3 : 1 [4]
white yellow green

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(ii) Test crosses were carried out on two white-fruited plants, **P** and **Q**, from the F2 generation. Each of these plants had its female flowers pollinated with pollen from a green-fruited plant.

For plant **P**, half of the offspring were white and half were yellow.
For plant **Q**, half of the offspring were white and half were green.

Deduce the genotypes of plants **P** and **Q**.

plant **P** Aa BB

plant **Q** Aabb

[2]

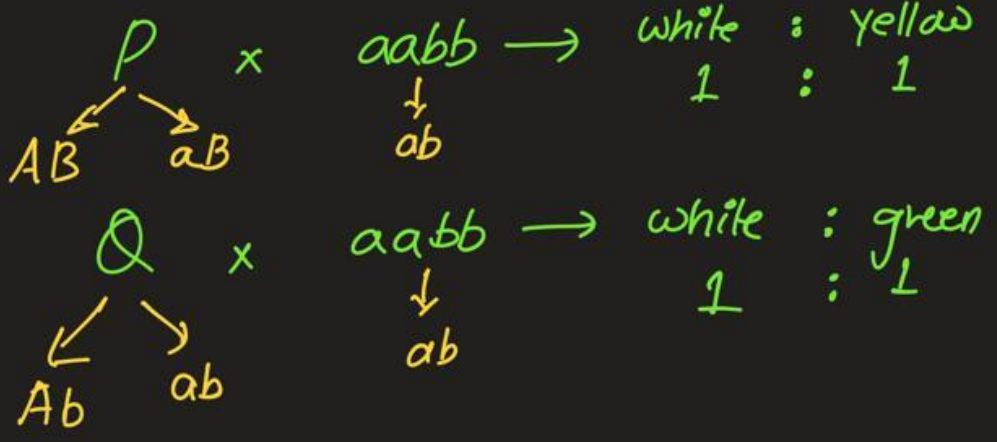
(iii) Plants **P** and **Q** show genetic variation with respect to fruit colour alleles.

Identify the process that occurred during meiosis in the F1 parents that produced this variation **and** the stage of meiosis at which it occurred.

process independent assortment

stage of meiosis metaphase I

[2]



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Interaction between gene loci
(epistasis)

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Interaction b/w gene loci (epistasis)

* Epistasis is the phenomenon of interaction b/w two or more non-linked genes at different gene loci to influence a given phenolype.

* We have discussed two examples of epistasis in genetic crosses done earlier (question 1 and question 2). The examples of fruit colour in patty pan squash plant and flower colour in morning glory plant were examples of epistasis.

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Q3.

2 The summer squash plant, *Cucurbita pepo*, produces edible fruits that vary in shape.
Fig. 2.1 shows the fruits of three different varieties of squash plants.

Patty pan (disc-shaped) | Di Nizza (spherical) | Alfresco (long)

Fig. 2.1

Fruit shape in squashes is controlled by two genes, **A/a** and **B/b**, that are located on different chromosomes.

- A disc-shaped fruit is produced when both dominant alleles, **A** and **B**, are present.
- A spherical fruit is produced when either allele **A** or allele **B** is present, but not if both **A** and **B** are present.
- A long fruit is produced when both allele **A** and allele **B** are absent.

(a) (i) Table 2.1 shows the possible genotypes of the Patty pan and Alfresco varieties.

Complete Table 2.1 to show the possible genotypes of the Di Nizza variety.

Table 2.1

variety	possible genotypes			
Patty pan (disc-shaped)	**AABB**	**AaBB**	**AABb**	**AaBb**
Di Nizza (spherical)	Aabb	AABb	aaBb	aaBB
Alfresco (long)	**aabb** (aabb)			

[1]

(ii) A gardener used pollen from a male flower of Alfresco to pollinate a female flower of Di Nizza. The gardener grew the seeds produced from this cross and found that half the offspring produced spherical fruits and half produced long fruits.

Draw **one** genetic diagram to explain this result.

parent genotypes: Aabb (spherical) × aabb (long)

gametes: Aabb → Ab (or aB), ab; aabb → ab

offspring genotypes: Aabb, aabb

offspring phenotypes: spherical : long

[4]

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(iii) The offspring show genetic variation with respect to fruit shape alleles.

Name the process that occurred during meiosis in the parents that produced this variation **and** state the stage of meiosis at which it occurred.

process ... independent assortment

stage of meiosis ... metaphase I ... [2]

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Q4.

(b) Flower colour is important in sexual reproduction of insect-pollinated plants.

In the rosy periwinkle, *Catharanthus roseus*, flower colour is controlled by three genes, **R/r**, **D/d** and **P/p**, which interact together to control flower colour.

Fig. 4.1 is a drawing of a rosy periwinkle.

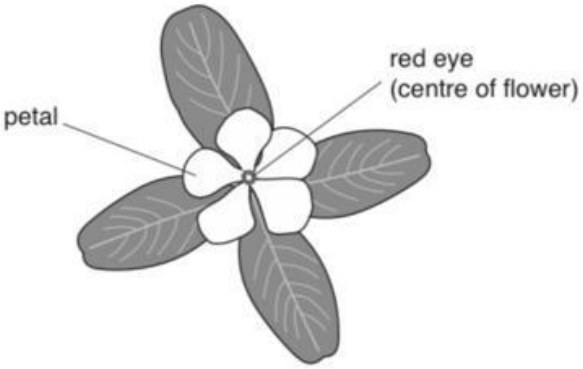

Fig. 4.1

The presence of the **R** allele results in a red pigment in the centre of the flower (red eye).

The **D** allele and the **P** allele are only expressed when the **R** allele is present.

- When the **D** allele and the **R** allele are present, the flower has dark pink petals with a red eye.
- When the **P** allele and the **R** allele are present, the flower has pale pink petals with a red eye.
- When the **D** allele, the **P** allele and the **R** allele are all present, the flower has dark pink petals with a red eye.
- The recessive alleles **r**, **d** and **p** result in no pigments being produced and the flower has white petals and no red eye.

(i) Deduce the phenotypes of these rosy periwinkle genotypes.

RR dd PP pale pink petals with red eye

Rr Dd Pp dark " " " " " "

rr Dd Pp white petals with no red eye

RR dd pp white petals with a red eye [4]

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Q5.

(b) Normal (wild-type) goats have a gold and black coat colour pattern, known as bezoar, and are also horned (have horns). Domestic goats may have a white coat and may be hornless (do not have horns).

These variations are coded for by two unlinked genes:

- white coat colour, coded for by the dominant allele of the gene **A/a**
- hornless, coded by the dominant allele of the gene **H/h**.

A cross between a white hornless goat and a bezoar horned goat produced offspring of four different phenotypes. aahh

Draw a genetic diagram to show the genotypes of the two parents, their gametes and the offspring, and the phenotypes of the offspring.

white, hornless — bezoar, horned

genotype of parents: Aa Hh × aahh

genotype of gametes: AH Ah aH ah — ah

offspring genotype: AaHh Aahh aaHh aahh

offspring phenotype: white, hornless; white, horned; bezoar, hornless; bezoar, horned

[4]

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The next question is on linkage. Before we attempt that, let's understand the significance of cross over value (COV).....

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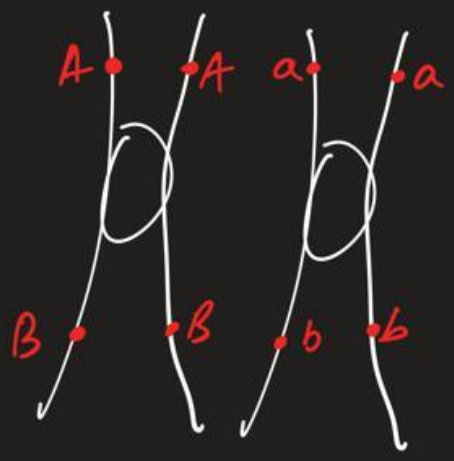
Significance of cross over value (COV)

- low COV
  - implies that the two genes are close together
- high COV
  - implies that the two genes are further apart



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Q6.

2 **(a)** In the sweet pea plant, *Lathyrus odoratus*, one gene codes for flower colour and one gene codes for pollen grain shape.
Flower colour is either purple or red. Pollen grain shape is either long or round.
The inheritance of these genes is an example of **autosomal linkage**.

- The allele **F** for purple flowers is dominant over the allele **f** for red flowers.
- The allele **G** for long pollen grains is dominant over allele **g** for round pollen grains.

Explain the meaning of the term *autosomal linkage*.

* the two genes are on the same pair of chromosomes
* the chromosomes are NOT sex chromosomes

[2]

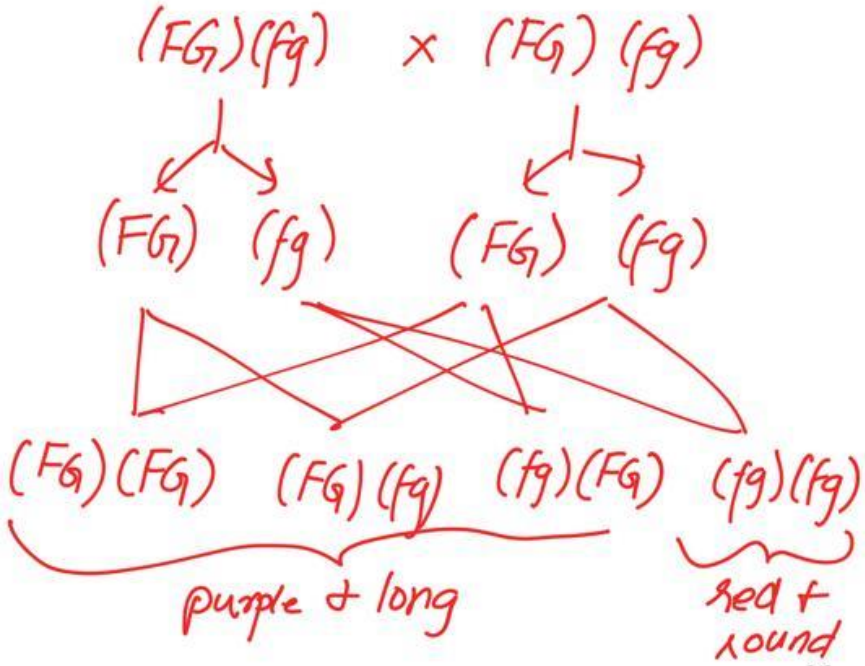
(FG)(FG) (fg)(fg)

(b) A dihybrid cross was carried out between homozygous dominant and homozygous recessive sweet pea plant parents to produce the F1 generation. (FG)(fg)

The offspring from the F1 generation were crossed to produce the F2 generation.

(i) Draw a genetic diagram to show a dihybrid cross between two offspring from the F1 generation.

Assume that these genes are closely linked and that there are **no** crossing over events.



[4]

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(ii) The actual results of the dihybrid cross are shown in Table 2.1.

Table 2.1

phenotypes of F2 offspring	number of individuals
purple flowers, long pollen grains	284
purple flowers, round pollen grains	21
red flowers, long pollen grains	21
red flowers, round pollen grains	55

State how the results support the fact that this is an example of autosomal linkage.

* phenotypic ratio is NOT 9:3:3:1

.....[1]

(c) (i) In a test cross, an individual of **known** genotype is crossed with an individual that has a dominant phenotype but unknown genotype.

State the genotype of the **known** individual in a test cross.

(fg) (fg)

.....[1]

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(ii) A test cross was carried out with sweet pea plants known to be heterozygous for both flower colour and pollen grain shape.

The results of the test cross are shown in Table 2.2.

Table 2.2

phenotypes of offspring of test cross	number of individuals
purple flowers, long pollen grains	215
purple flowers, round pollen grains	30
red flowers, long pollen grains	32
red flowers, round pollen grains	210

The result of a test cross can be used to determine a crossover value (COV).
A crossover value is the percentage of the total number of offspring showing recombination.

The crossover value (COV) can be calculated using the formula shown in Fig. 2.1.

$$\text{COV} = \frac{\text{number of recombinants}}{\text{total number of individuals}} \times 100$$

Fig. 2.1

Calculate the COV from the results shown in Table 2.2.

$COV = 62/487 \times 100$

COV = 12.7 % [1]

(iii) Suggest what information about the relative distance between the linked genes can be gained from crossover values.

low COV implies that the two genes are close together. [1]

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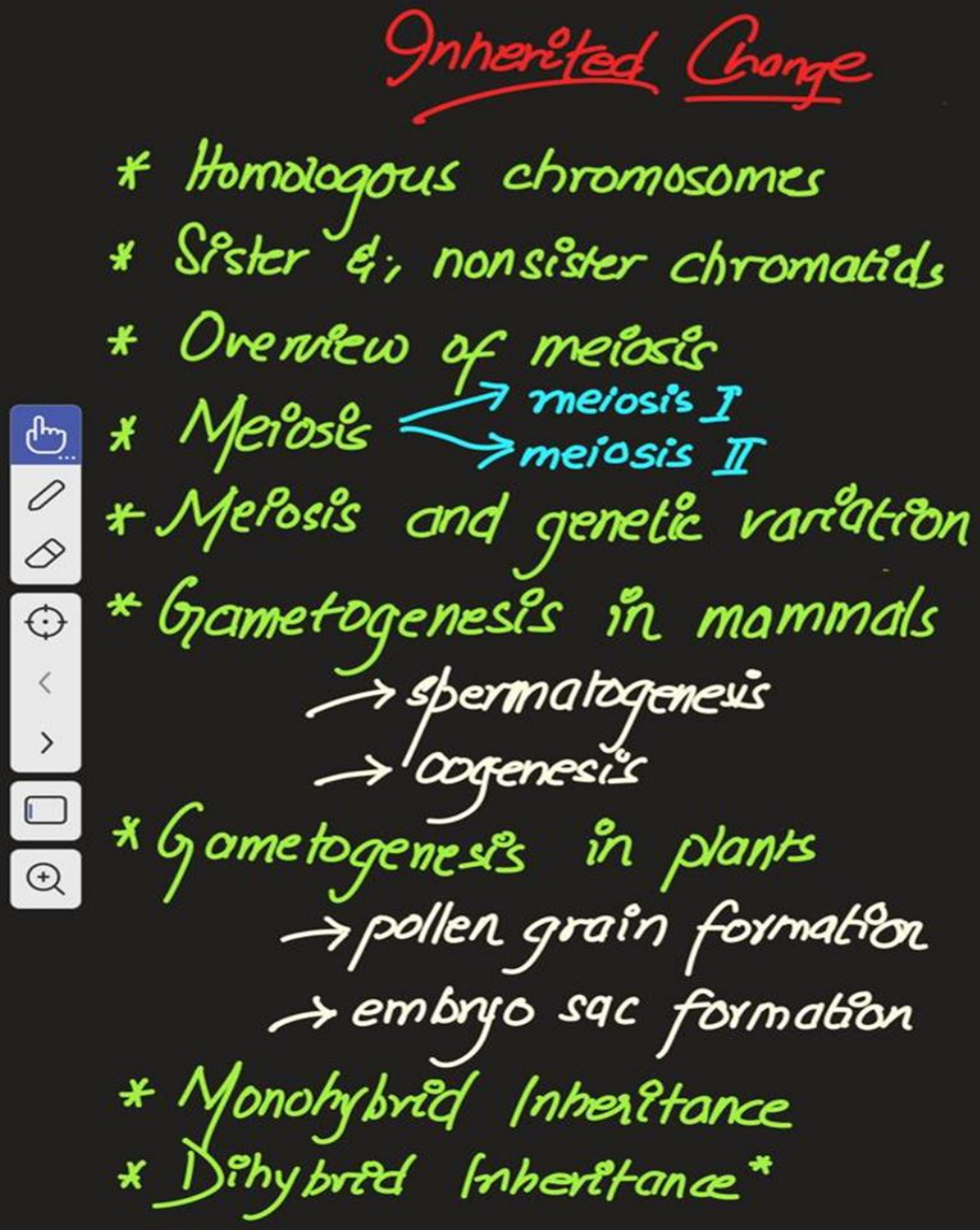

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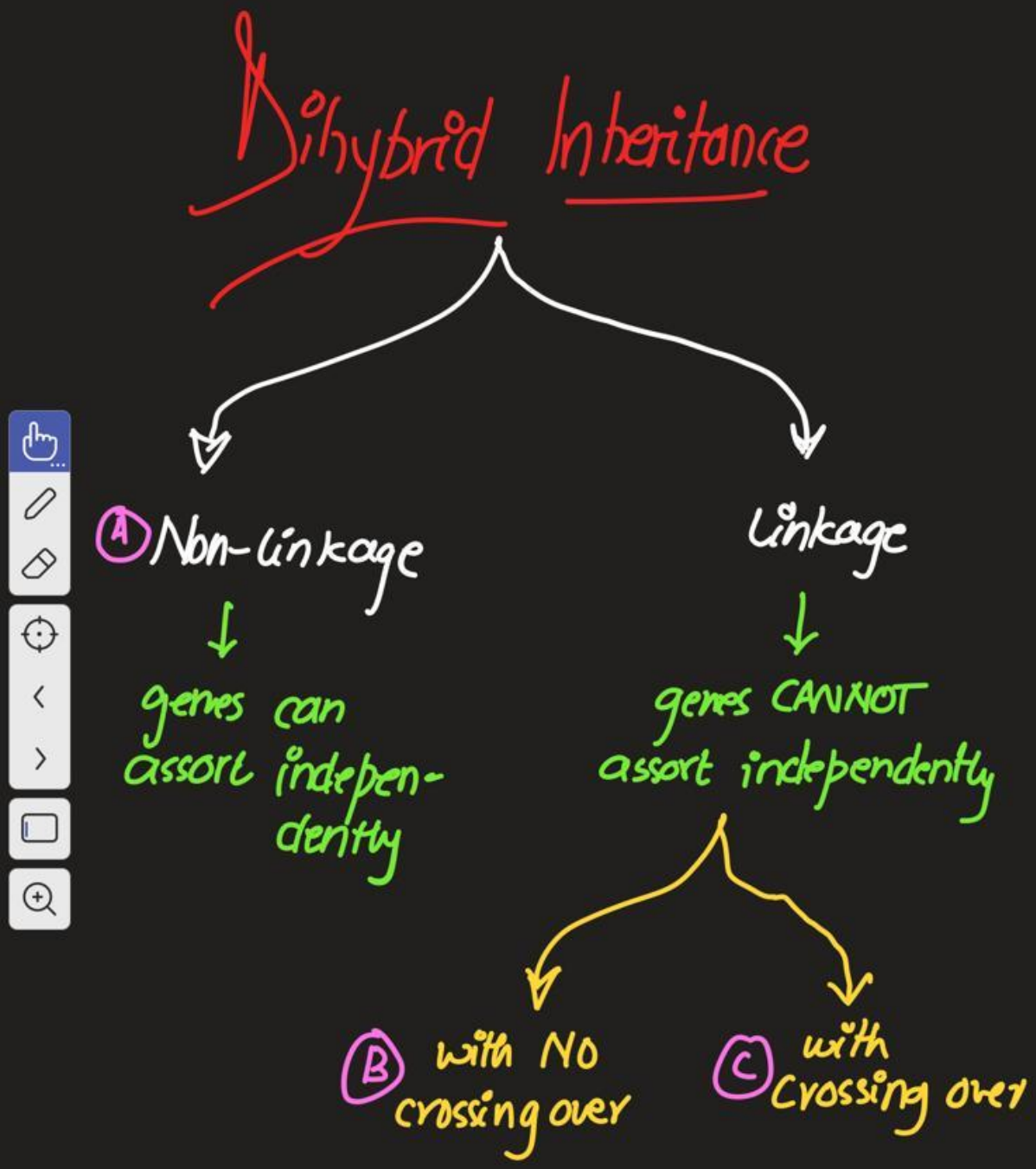
Dihybrid Inheritance
A Non-linkage
genes can assort indepen-dently
Linkage
genes CANNOT assort independently
B with No crossing over
C with crossing over

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Q.

5 Fig. 5.1 shows a cat displaying the recessive phenotype for two unlinked genes.

- **H/h** controls hair length. The allele for short hair is dominant to the allele for long hair.
- **R/r** controls coat pattern. The allele for 'full colour', with pigment on all parts of the body, is dominant to the allele for 'pointed', where the pigment is restricted to the ears, face, paws and tail.

hhrr

Fig. 5.1

(a) Draw a genetic diagram to predict the offspring genotypes and phenotypes when the cat in Fig. 5.1 is crossed with a cat that is heterozygous for the hair length gene and heterozygous for the coat pattern gene.

parent phenotypes: short hair, full colour × long hair, pointed

parent genotypes: HhRr × hhrr

gametes: HR Hr hR hr ; hr

F1 genotypes: HhRr Hhrr hhRr hhrr

F1 phenotypes: short hair, full color ; short hair, pointed ; long hair, full color ; long hair, pointed

[5]

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Q.

5 Scallops, which are bivalve molluscs, are important commercially throughout the world. The marine bay scallop, *Agropecten irradians*, has three distinct shell colours, yellow, orange and black. The shell colour is controlled by a gene with three alleles, yellow, S^y, orange, S^o, and black, S^b.

Scallops are hermaphrodite and are able to fertilise themselves to produce offspring.

Single mature adult specimens of yellow, orange and black scallops were collected and kept in separate tanks of seawater until they produced young. The young were then scored for shell colour. The results were as follows.

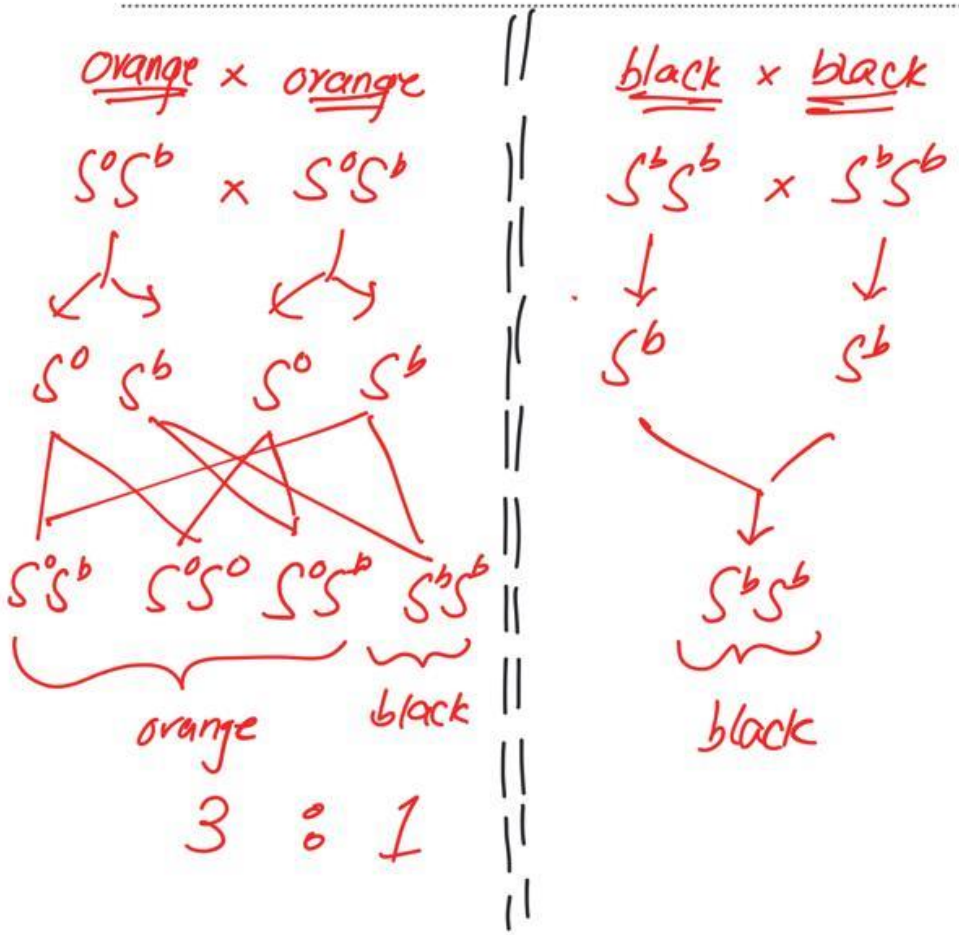
yellow scallop – 25 yellow and 8 black ✗
orange scallop – 31 orange and 9 black ✓
black scallop – 27 black ✓

(a) Explain the results from the orange and black scallops, using the symbols given.

.....

.....

.....



[6]

HEUSER

Q.

7 The fruit fly, *Drosophila melanogaster*, has many phenotypic variations and has been used in experiments to demonstrate the principles of inheritance.

(a) The majority of fruit flies have red eyes but there is a variant with white eyes.

Fig. 7.1 shows the red-eyed and white-eyed variants of the fruit fly.

Fig. 7.1

The gene for eye colour is located on the X chromosome.

Using suitable symbols, draw a genetic diagram to show the possible offspring of a cross between a heterozygous red-eyed female fruit fly with a white-eyed male fruit fly.

key to symbols:

X^R → red allele

X^r → white allele

parental phenotypes	red-eyed female	white-eyed male
parental genotypes	X^RX^r	X^rY
gametes	X^R X^r	X^r Y
offspring genotypes	X^RX^r	X^RY X^rX^r X^rY
offspring phenotypes	red-eyed female	red-eyed male white eyed female white eyed male

[5]

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(b) One of the genes controlling the clotting of blood in humans is also located on the X chromosome. A rare variation of the gene, a recessive allele for haemophilia, can lead to a condition where the blood fails to clot properly.

(i) State why a man who has haemophilia is unable to pass the condition on to his son.

s/c a man passes on the Y chromosome to his son

.....[1]

(ii) Queen Victoria of Great Britain in the 19th century was a carrier of haemophilia, but did not have the condition.

State the term used to describe the genotype of a carrier.

heterozygous

.....[1]

(iii) Neither of Queen Victoria's parents carried the allele for haemophilia.

Suggest how Queen Victoria could have become a carrier.

* random mutation

.....[1]

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Q.

7 **(a)** The stems of raspberry plants have spines.

Fig. 7.1 shows part of a raspberry plant.

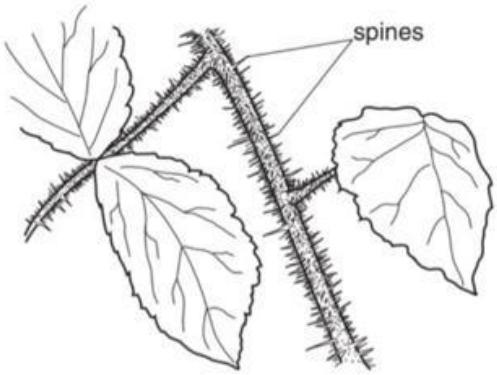

Fig. 7.1

The colour of the spines is controlled by two genes, **A/a** and **B/b**. The two genes are on different pairs of chromosomes.

- Allele **A** produces a pink anthocyanin pigment in the spines.
- Allele **B** has no effect by itself, but increases the colour produced by allele **A** to give red spines.
- Alleles **a** and **b** have no effect on colour.
- In the absence of anthocyanin, the spines are green.

State the colour of the spines of raspberry plants with the genotypes **Aabb** and **aaBB**.

Aabb pink

aaBB green [2]

(b) Plants with the genotype **AaBb** were crossed with plants with the genotype **aabb**. The resulting seeds were sown and the seedlings grown until their stems developed spines.

Use a genetic diagram to show the outcome of this cross, including the ratio of offspring phenotypes.

parental phenotype: red × green

parental genotype: AaBb × aabb

meiosis

gametes: AB Ab aB ab | ab

offspring genot.: AaBb Aabb aaBb aabb

offspring phenot.: red pink green

ratio: 1 : 1 : 2

[5]

(c) Suggest why the ratio you have given in your genetic diagram would be different if the genes **A/a** and **B/b** were on the same homologous pair of chromosomes.

* the two genes will be inherited together
* with no independent assortment
* there will be only two phenotypes

[2]

HEUSER

Q.

7 **(a)** Explain what is meant by the term *heterozygous genotype*.

heterozygous ..... different alleles of a gene .....

.....

genotype ..... genetic constitution of an organism .....

.....[2]

(b) The budgerigar, *Melopsittacus undulatus*, is a small type of parrot that is native to Australia.

Fig. 7.1 shows a budgerigar.

Fig. 7.1

A budgerigar can have blue, green, yellow or white feathers.

Two genes, **A**/**a** and **D**/**d**, are involved in the inheritance of feather colour in budgerigars.

- A bird which has at least one dominant allele **A** but is homozygous for **d** has blue feathers. Aadd AAdd
- A bird which has at least one dominant allele **D** but is homozygous for **a** has yellow feathers. aaDd aaDD
- A bird with at least one dominant **A** allele **and** one dominant **D** allele has green feathers. AaDd AADd AaDD AADD
- A bird that is homozygous for **a** and **d** has white feathers.

aadd

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(c) Two green-feathered budgerigars, heterozygous at both gene loci, were crossed.

Draw a genetic diagram of this cross to show the probability of producing offspring with yellow feathers.

parental genotypes → AaDd × AaDd

genotype of gametes

	AD	Ad	aD	ad
AD	AADD green	AADd green	AaDD green	AaDd green
Ad	AADd green	AAdd blue	AaDd green	Aadd blue
aD	AaDD green	AaDd green	aaDD* yellow	aaDd* yellow
ad	AaDd green	Aadd blue	aaDd* yellow	aadd white

probability of yellow feathers = 3/16

[6]

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X^2 -test

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X^2-test

* In pea plants, yellow colour of the seeds is dominant to green colour. Seed colour is determined by a gene with two alleles:

R → yellow

r → green

Pure-breeding pea plants with yellow (RR) seeds were crossed with pure-breeding pea plants with green (rr) seeds. All the offsprings had yellow (Rr) seeds. These seeds were grown and the resultant plants were allowed to self-pollinate yielding 600 offsprings with the following characteristics:

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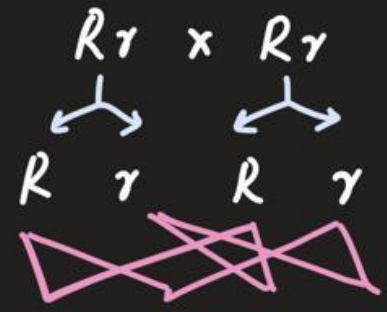
405 yellow seeds
195 green seeds

* We will refer to these results as the 'observed' results.

Q: What were our 'expected results';

Ans: We expected the offsprings of heterozygous parents to be in the ratio 3:1.



Yellow : green

3 : 1

E ⇒ 450 : 150

O ⇒ 405 : 195

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* In the case of 600 offsprings, our 'expected' numbers would have been:

	Yellow	Green
Expected (E)	450	150
Observed (O)	405	195

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Q: Are the 'OBSERVED' numbers SIGNIFICANTLY DIFFERENT from our 'EXPECTED' numbers? OR is the difference just DUE TO CHANCE?

* X^2-test can help us determine that !!!
* X^2- test is used to determine the X^2-value which can help us answer if there is a significant difference b/w the observed and expected numbers.

* Before we calculate the X^2-value, let's make a NULL HYPOTHESIS =>

"There is no significant difference b/w

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The observed$_{(O)}$ and expected$_{(E)}$ numbers'

① We will 'reject the null hypothesis' if the X^2- value ascertains there is a significant difference.

② We will 'accept the null hypothesis' if the X^2- value ascertains there is no significant difference. The difference is due to chance.

• Now, let's determine the X^2- value:

$$X^2 = \sum \frac{(O-E)^2}{E}$$

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* for our case,

	Yellow	Green
O	405	195
E	450	150
(O-E)	-45	+45
$(O-E)^2$	2025	2025
$(O-E)^2/E$	4.5	13.5

$$X^2 = \Sigma(4.5 + 13.5) = 18 //$$

Q: What's the next step?

Ans: Determine the degrees of freedom (v) for this case.

(v) degrees of freedom = $n - 1$

- where n = no. of categories

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- The different phenotypes are the categories in this case.

Therefore, $v = 2 - 1 = 1$ //

* Now we have the → X^2 - value = 18
  → $v = 1$

* We will now move to the X^2- table for our final answer.

Q: What is a X^2-table?

Ans: It shows the distribution of X^2- values for various degrees of freedom and p-values

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Q: What is a p-value?

Ans: p-value is the probability that the "null hypothesis is correct". In biological experiments, the null hypothesis is rejected ONLY if the p-value is LESS THAN 0.05!!!

① if the calculated X^2-value corresponds to $p < 0.05$ → the null hypothesis is rejected. There is a significant difference b/w the observed and expected results. The difference in results is NOT due to chance.

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③ if the calculated X^2-value corresponds to $p \geq 0.05$ → the null hypothesis is accepted. There is no significant difference b/w the observed and expected results. The difference in results is just due to chance.

* X^2- test is generally used in genetics & ecological sampling.

* For the X^2- test to be used;

1) The data must be discrete (categorical)
2) The sample size should be greater than 20.

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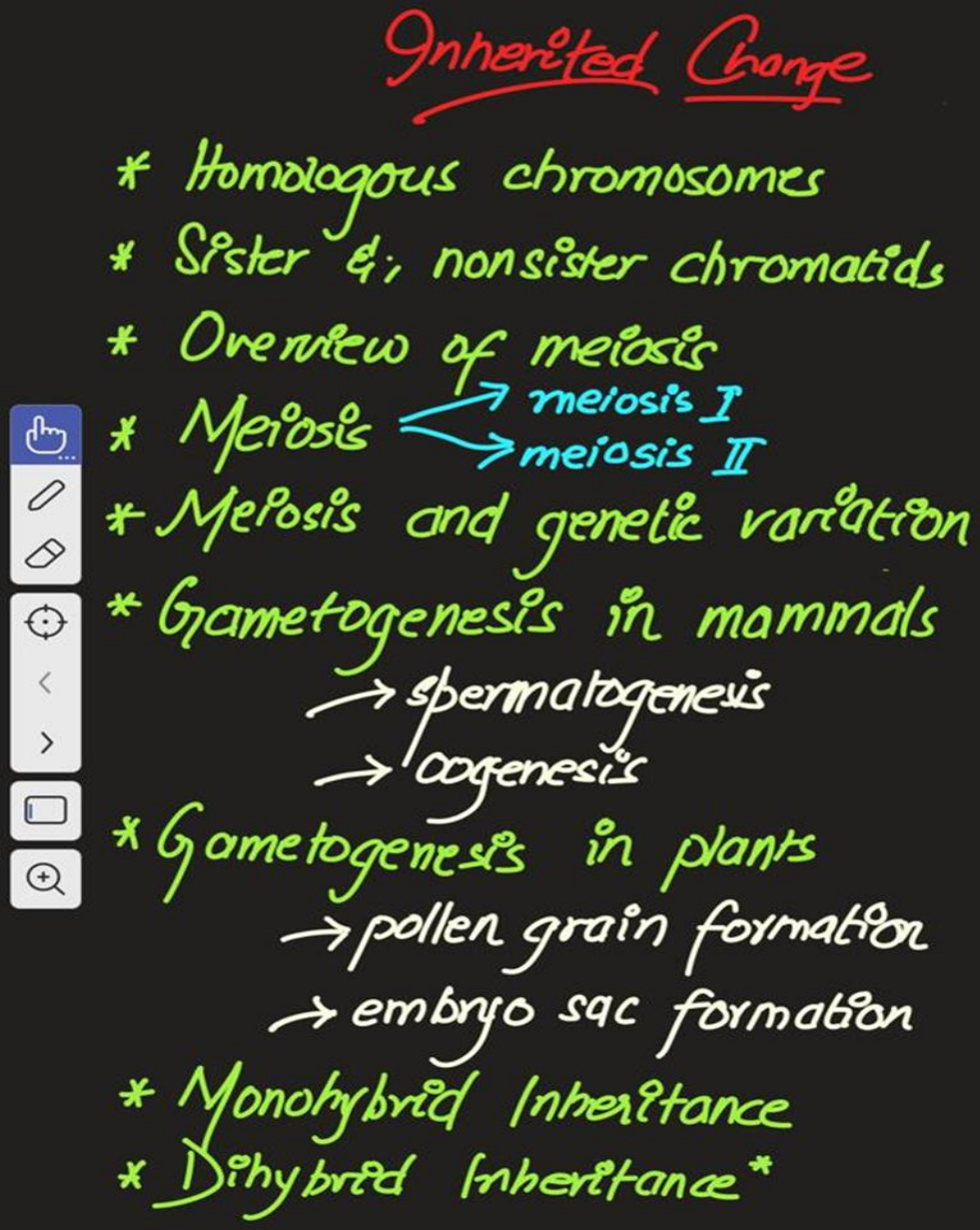

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Inherited Change

X^2 - test

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Previously....

$X^2 = 18$
$df = 1$

critical values

Table/Critical values of χ^2

Degrees of freedom	Probability										
	0.95	0.90	0.80	0.70	0.50	0.30	0.20	0.10	0.05	0.01	0.001
1	0.004	0.02	0.06	0.15	0.46	1.07	1.64	2.71	3.84	6.64	10.83
2	0.10	0.21	0.45	0.71	1.39	2.41	3.22	4.60	5.99	9.21	13.82
3	0.35	0.58	1.01	1.42	2.37	3.66	4.64	6.25	7.82	11.34	16.27
4	0.71	1.06	1.65	2.20	3.36	4.88	5.99	7.78	9.49	13.28	18.47
5	1.14	1.61	2.34	3.00	4.35	6.06	7.29	9.24	11.07	15.09	20.52
6	1.63	2.20	3.07	3.83	5.35	7.23	8.56	10.64	12.59	16.81	22.46
7	2.17	2.83	3.82	4.67	6.35	8.38	9.80	12.02	14.07	18.48	24.32
8	2.73	3.49	4.59	5.53	7.34	9.52	11.03	13.36	15.51	20.09	26.12
9	3.32	4.17	5.38	6.39	8.34	10.66	12.24	14.68	16.92	21.67	27.88
10	3.94	4.86	6.18	7.27	9.34	11.78	13.44	15.99	18.31	23.21	29.59
	Nonsignificant								Significant		

X^2-value → corresponds to $p < 0.001$

↓

* reject the null hypothesis
* there is a significant difference between obs. & exp. results.

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X^2 - test

Q1. WHEN ??

Ans * to find out if there is a significant difference bw the observed (O) and the expected (E) results
* when the data is discrete

Q2. HOW ??

Ans
1. Make a null hypothesis
2. Calculate the χ^2-value
3. Determine the degree of freedom (df)
4. Consult the χ^2 table

If the X^2 value is GREATER than the critical value at $p=0.05$

It implies that $p<0.05$,
→ reject the null hypothesis
→ there is a significant diff bw the O and E results
→ difference in results is NOT due to chance

If the X^2 value is LESSER than or EQUAL to the critical value at $p=0.05$

It implies that $p \geq 0.05$
→ accept the null hypothesis
→ NO significant diff. bw O and E results
→ diff. in results is due to chance

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Q1.

6 The fruit fly, *Drosophila melanogaster*, has eyes, a striped abdomen and wings longer than its abdomen. This is called a 'wild-type' fly.

Mutation has resulted in many variations of these features.

Table 6.1 shows diagrams of a wild-type fly and three other flies, each of which shows **one** recessive mutation.

Table 6.1

	'wild'			
eyes	present	present	absent recessive	present
abdomen	striped	black recessive	striped	striped
wing description	long	long	long	short recessive

(a) Using appropriate symbols, complete the genetic diagram below.

symbols

E → with eyes

e → no eyes

A → striped

a → black

parental phenotypes	with eyes black abdomen (dom. / rec.)	X	no eyes striped abdomen (rec. / dom.)
parental genotypes	Ee aa		ee Aa
gametes	Ea ea		eA ea
offspring genotypes	Eeaa	eeaa	EeAa	eeAa
offspring phenotypes	with eyes black abdomen	no eyes black abdomen	with eyes striped abdomen	no eyes striped abdomen

[4]

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(b) State how you would carry out a test cross.

cross it with a homozygous recessive organism (no eyes, black abdomen, short wings) [1]

(c) A cross was carried out between a fly heterozygous for striped abdomen and long wings and a fly with a black abdomen and short wings. (aaww) (AaWw)

The results are shown below in Table 6.2.

Table 6.2

	offspring	number
1	striped abdomen long wing	86
2	black abdomen long wing	87
3	striped abdomen short wing	81
4	black abdomen short wing	78
	total	332

AaWw aaww → aw

AW
Aw
aW
aw

A chi-squared test (χ^2) was carried out on these data.

Complete Table 6.3 **and** calculate the value of χ^2.

Table 6.3

observed number (O)	expected number (E)	O – E	$(O - E)^2$	$\frac{(O - E)^2}{E}$
86	83	3	9	0.11
87	83	4	16	0.19
81	83	-2	4	0.05
78	83	-5	25	0.30
332	332			

$$\chi^2 = \sum \frac{(O - E)^2}{E}$$

$\sum$ = sum of...

df = 4-1 = 3

χ^2 0.65 [3]

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(d) Table 6.4 shows χ^2 values.

$\chi^2 = 0.65$

Table 6.4

degrees of freedom	probability						
	0.50	0.20	0.10	0.05	0.02	0.01	0.001
3	2.37	4.64	6.25	7.82	9.84	11.34	16.27

Using Table 6.4, explain what conclusions can be made about the results of the χ^2 test.

* χ^2 value is greater than the critical value (7.84) which corresponds to $p > 0.05$.
* This implies that there is no significant diff between the observed and expected results.

[2]

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Q2.

2 A maize plant produced a total of 381 grains, 216 purple and smooth, 79 purple and shrunken, 65 yellow and smooth and 21 yellow and shrunken.

(a) Using the symbols **A** for purple and **a** for yellow and **B** for smooth and **b** for shrunken, draw a genetic diagram to explain these results.

parental genotype: AaBb x AaBb

parental gametes: AB Ab aB ab

	AB	Ab	aB	ab
AB				
Ab				
aB				
ab				

[4]

(b) Explain why yellow shrunken grains breed true.

b/c it is homozygous recessive for both the genes (aabb)

[2]

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A chi-squared test was carried out to test the significance of the differences between the observed and expected results.

Table 2.1

grain phenotype	observed number	observed ratio	expected ratio	expected number	[obs no. – exp no.]^2 ÷ expected no.
purple and smooth	216	10.3	9	381 × 9/16 = 214	4/214 = 0.019
purple and shrunken	79	3.8	3	381 × 3/16 = 71	64/71 = 0.901
yellow and smooth	65	3.1	3	381 x 3/16 = 71	0.507
yellow and shrunken	21	1.0	1	381 x 1/16 = 24	0.375
total number	381			chi square value	1.80

(c) Complete the missing spaces in the Table 2.1 [3]

Table 2.2

	probability greater than						
degrees of freedom	0.50	0.20	0.10	0.05	0.02	0.01	0.001
3	2.37	4.64	6.25	7.82	9.84	11.34	16.27

(d) Use the calculated values of chi-squared test and the table of probabilities to find the probability of the observed ratio of phenotypes differing significantly from the expected.

P > 0.50 [1]

(e) State what conclusions may be drawn from the probability found in **(d)**.

* no significant diff bw observed and expected results b/c the calculated X^2 value is lesser than the critical value (7.82). [2]

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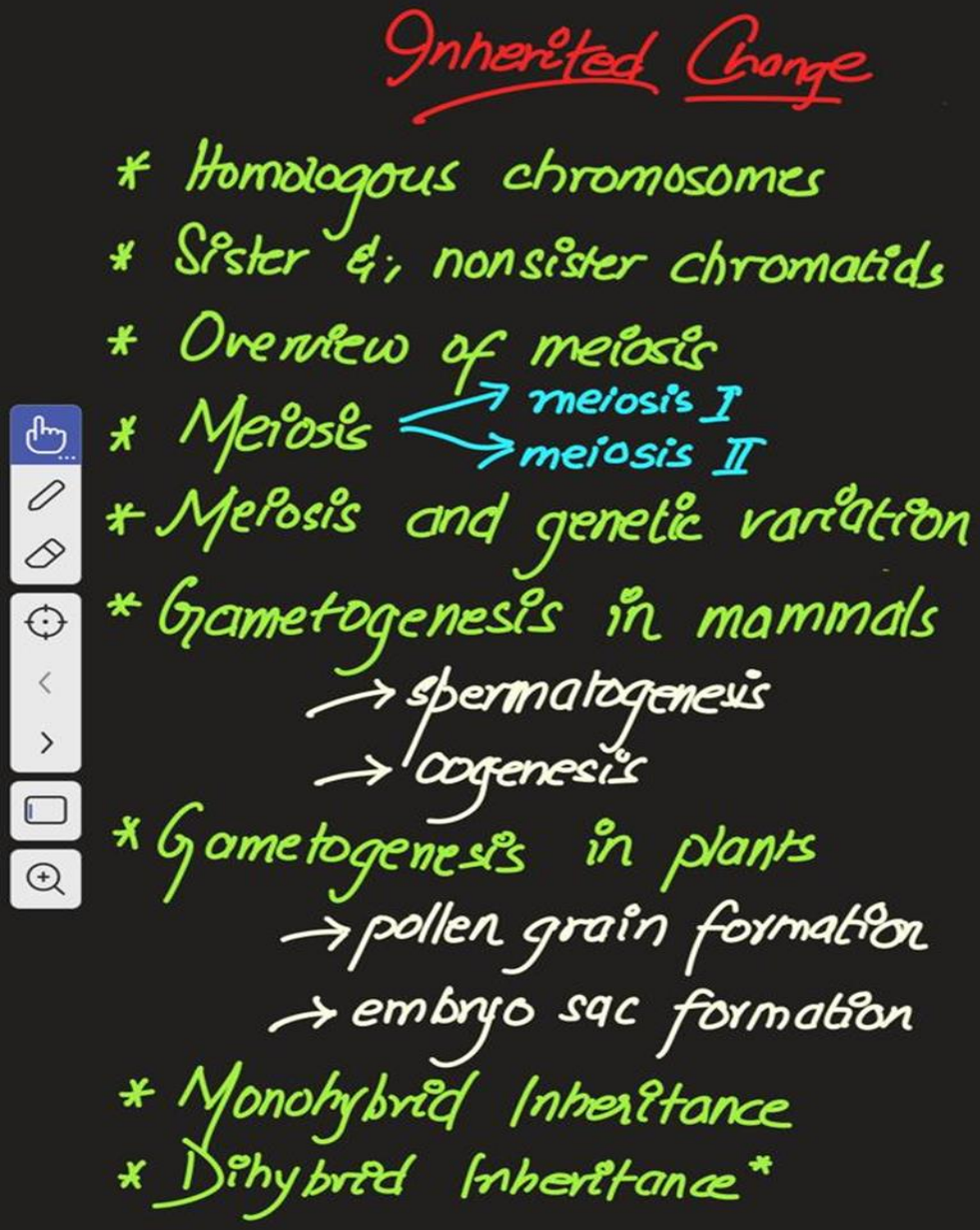

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X^2 - test

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X^2 - test

Q1. WHEN ??

Ans * to find out if there is a significant difference bw the observed (O) and the expected (E) results
* when the data is discrete

Q2. HOW ??

Ans
1. Make a null hypothesis
2. Calculate the X^2-value
3. Determine the degree of freedom (df)
4. Consult the X^2 table

If the X^2 value is GREATER than the critical value at $p=0.05$

It implies that $p<0.05$,
→ reject the null hypothesis
→ there is a significant diff bw the O and E results
→ difference in results is NOT due to chance

If the X^2 value is LESSER than or EQUAL to the critical value at $p=0.05$

It implies that $p \geq 0.05$
→ accept the null hypothesis
→ NO significant diff. bw O and E results
→ diff. in results is due to chance

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Q3.

2 Melanism (dark-coloured wings) in a species of moth is controlled by a gene with two alleles. The dominant allele codes for the melanic form and the recessive allele codes for the non-melanic form (pale, speckled wings). Birds are a predator of this species.

The melanic form is better camouflaged in areas polluted by carbon deposits from smoke. The non-melanic form is better camouflaged in areas unpolluted by smoke.

Mm

(e) The investigators carried out test crosses by breeding a heterozygous male melanic moth with several female non-melanic moths. The number of moths of each form in the next generation was recorded.

mm

melanic	56
non-melanic	48

Mm × mm

M m m m

Mm : mm

(i) Complete Table 2.1 to calculate the chi-squared (χ^2) value for determining whether or not these results were significantly different from those expected for crosses of this type.

The formula for calculating χ^2 is:

$$\chi^2 = \Sigma \frac{(O-E)^2}{E} \qquad v = c - 1$$

v = degrees of freedom $\qquad$ c = number of classes

Table 2.1

category	O	E	$\frac{(O-E)^2}{E}$
melanic	56	52	0·31
non-melanic	48	52	0·31
		$\chi^2 =$	0·62

[3]

(ii) Table 2.2 shows some critical values for χ^2 at different probabilities.

Table 2.2

degrees of freedom	probability						
	0.99	0.95	0.90	0.10	0.05	0.01	0.001
1	0.0002	0.0039	0.0158	2.706	3.841	6.635	10.827
2	0.0201	0.1026	0.2107	4.605	5.991	9.210	13.815

Use Table 2.2 to explain whether or not the χ^2 value calculated in Table 2.1 is significant.

X^2 value corresponds to $p > 0.05$ which implies that there is no significant difference bw observed and expected results.

[1]

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Q4. (ecological sampling)

2 Cereal crops are often sprayed with selective herbicides which can reduce the population of the local wildlife. One method of helping to conserve wildlife is to leave a 6 m strip, called a headland, around fields where cereal crops are grown. The headland is not sprayed with any herbicides.

An investigation was carried out into the effect on the butterfly populations of leaving headlands unsprayed.

- Two groups of 20 fields growing the same cereal crop were studied.
- The headlands of one group of 20 fields were left unsprayed by herbicide.
- The headlands of the other group of 20 fields were sprayed with herbicide.
- The total number of each species of butterfly was counted in each group of 20 fields.
- A chi-squared (χ^2) test was used to find out if the differences in the butterfly populations were significant.

(a) State two variables that were standardised in this investigation.

1 number of fields studied

2 width of the headland [1]

(b) **(i)** State a reason why the chi-squared (χ^2) test is suitable to use for comparing the butterfly populations.

to determine the diff. bw observed and expected results [1]

(ii) State a null hypothesis for the chi-squared test for this investigation.

no significant difference bw the no. of butterflies of each species when headland sprayed and when not sprayed [1]

(iii) Table 2.1 shows the results for one of the species of butterfly, species **Q**.

Complete Table 2.1 to calculate the value of χ^2 using the equation below.

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

$$v = c - 1$$

O = observed result
E = expected result
v = degrees of freedom
c = number of classes

Table 2.1

species **Q**	O	E	$(O-E)^2$	$\frac{(O-E)^2}{E}$
number on headland sprayed with herbicide	3	20	289	14.45
number on headland not sprayed with herbicide	37	20	289	14.45

χ^2 = 28.9 [3]

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Table 2.2 shows some chi-squared (χ^2) values.

Table 2.2

degrees of freedom	p <			
	0.10	0.05	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.61	5.99	9.21	13.82
3	6.25	7.82	11.34	16.27

< 0.001
↑
28.9

(iv) State the critical value at $p < 0.05$ for this chi-squared test. 3.84 [1]

(v) State what you conclude from the result of the chi-squared test for species **Q**.

there is a significant difference in the butterfly population [1]

due to herbicide spray

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Q5. (ecological sampling)

3 (a) Grassland, grazed by goats, was compared to grassland ungrazed for varying amounts of time. Moths were trapped, their numbers counted and then the moths were released. The effect of grazing on a species of rare moth was investigated. A chi-squared (χ^2) test was used to test the significance of the results obtained.

(i) State the null hypothesis.

r:o significant difference in the population of moth in grazed and ungrazed land (for varying amount of time) [1]

(ii) Complete Table 3.1 to calculate the value of χ^2 from the equation below.

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$ O = Observed result, E = Expected result

Table 3.1

site	O	E	$(O-E)^2$	$\frac{(O-E)^2}{E}$
grazed for 2 years	36	80	1936	24.2
ungrazed for 10 years	90	88	100	1.25
ungrazed for 30 years	114	80	1156	14.45

χ^2 = 39.9 [4]

Table 3.2 shows some chi-squared values.

Table 3.2

degrees of freedom	probability greater than			
	0.10	0.05 →	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.60	5.99 →	9.21 →	13.82 → 39.9
3	6.25	7.82	11.34	16.27
4	7.78	9.49	13.28	18.46
10	15.99	18.31	23.21	29.59
20	28.40	31.41	37.57	45.31

$p < 0.001$

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(b) (i) State the number of degrees of freedom. 2 [1]

(ii) State the probability of the value calculated for χ^2. $p < 0.001$ [1]

(iii) Explain the significance of these results and suggest the consequences to this species of moth if grazing land is increased.

grazing has a significant effect on the population of moth. Increasing the grazed land will decrease the moth population due to habitat destruction

[3]

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Q6.

7 (a) The fruit fly, *Drosophila melanogaster*, feeds on sugars found in damaged fruits. A fly with normal features is called a wild type. It has a striped body and its wings are longer than its abdomen. There are mutant variations such as an ebony coloured body or vestigial wings. These three types of fly are shown in Fig. 7.1.

wild type ebony body vestigial wing

A → striped body
a → ebony bodied
B → long wings
b → short wings

Fig. 7.1

Wild type features are coded for by dominant alleles, **A** for wild type body and **B** for wild type wings.

Explain what is meant by the terms *allele* and *dominant*.

allele ..alternative forms of a gene..

dominant ..an allele is said to be dominant if it expresses itself in the homozygote and the heterozygote.. [2]

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(b) Two wild type fruit flies were crossed. Each had alleles **A** and **B** and carried alleles for ebony body and vestigial wings.

Draw a genetic diagram to show the possible offspring of this cross.

parental genotype AaBb × AaBb

gamete AB, Ab, aB, ab

	AB	Ab	aB	ab
AB				
Ab				
aB				
ab				

9 : 3 : 3 : 1

[6]

(c) When the two heterozygous fruit flies in **(b)** were crossed, 384 eggs hatched and developed into adult flies.

A chi-squared (χ^2) test was carried out to test the significance of the differences between observed and expected results.

$$\chi^2 = \Sigma \frac{(O - E)^2}{E}$$

where Σ = sum of
O = observed value
E = expected value

(i) Complete the missing values in Table 7.1.

Table 7.1

	phenotypes of *Drosophila melanogaster*			
	grey body long wing	grey body vestigial wing	ebony body long wing	ebony body vestigial wing
observed number (O)	207	79	68	30
expected ratio	9	3	3	1
expected number (E)	216	72	72	24
O – E	-9	7	-4	6
$(O - E)^2$	81	49	16	36
$\frac{(O - E)^2}{E}$	0.38	0.68	0.22	1.50

[3]

(ii) Calculate the value for χ^2.

χ^2 = 2.78 [1]

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Table 7.2 relates χ^2 values to probability values.

As four classes of data were counted the number of degrees of freedom was 4 − 1 = 3. Table 7.2 gives values of χ^2 where there are three degrees of freedom.

Table 7.2

probability greater than	*0.50*	*0.20*	*0.10*	*0.05*	*0.01*	*0.001*
values for χ^2	2.37	4.64	6.25	7.82	11.34	16.27

2·78

(iii) Using your value for χ^2, and Table 7.2, explain whether or not the observed results were significantly different from the expected results.

X^2 value corresponds to $p > 0.05$ which implies that there is no significant diff. bw observed and expected numbers.

.....[2]

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Gene Control

* controlling the expression of a gene
  transcription + translation

* is achieved via transcription factors (proteins)

* GENES → Structural genes
  → Regulatory genes
  ↓
  code for proteins which control the expression of genes

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Gene Control

① Gene control refers to the control of gene expression in cells.

② Gene control is essential so that proteins are produced in appropriate quantities at appropriate times in a specific cell.

③ Transcription factors are proteins which control gene expression.

④ The larger the genome, the greater the number of genes which code for transcription factors.

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⑤ Genes can be classified into the following two types:

A) Structural genes, which code for proteins that are needed for a functional role within the cell, for e.g. enzymes & hormones

B) Regulatory genes, which code for proteins that regulate the expression of the other genes (such as transcription factors)

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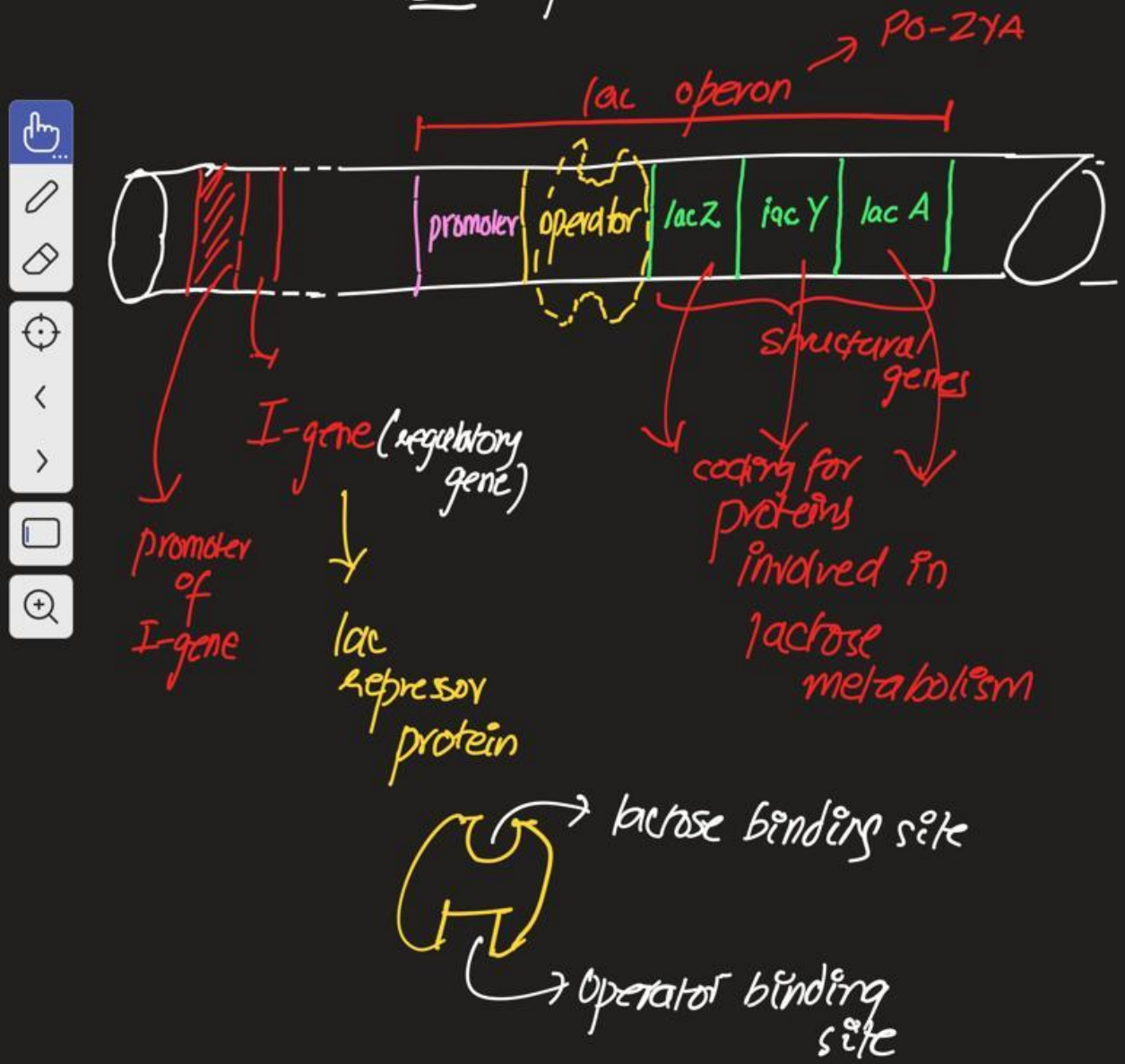
Gene Control in Prokaryotes

⇒ Lac Operon

(lactose operon)



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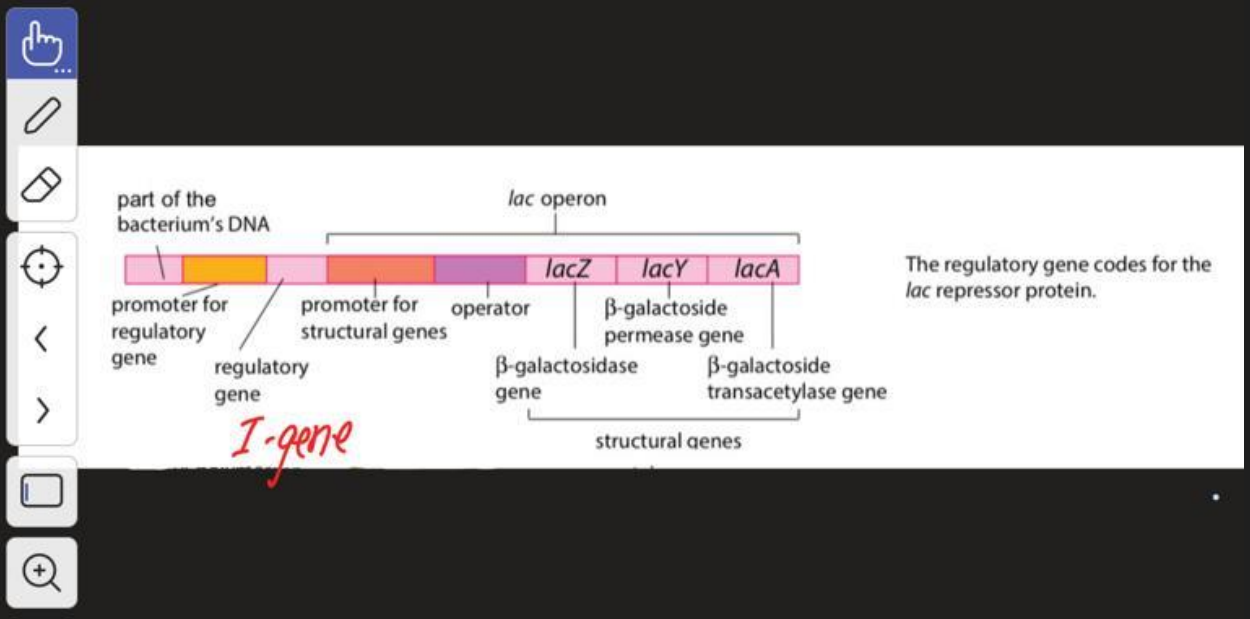
part of the bacterium's DNA
lac operon
lacZ
lacY
lacA
promoter for regulatory gene
regulatory gene
promoter for structural genes
operator
β-galactosidase gene
β-galactoside permease gene
β-galactoside transacetylase gene
structural genes
The regulatory gene codes for the lac repressor protein.
I-gene

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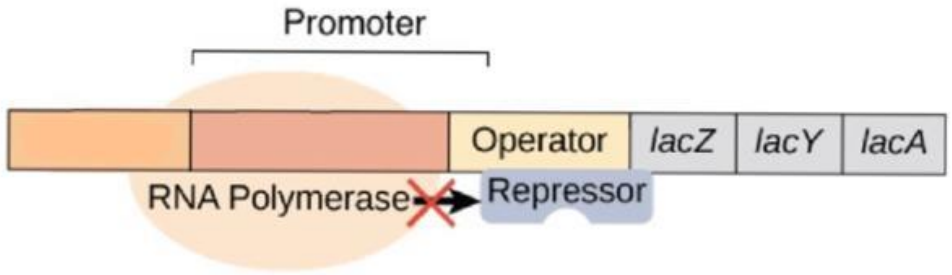
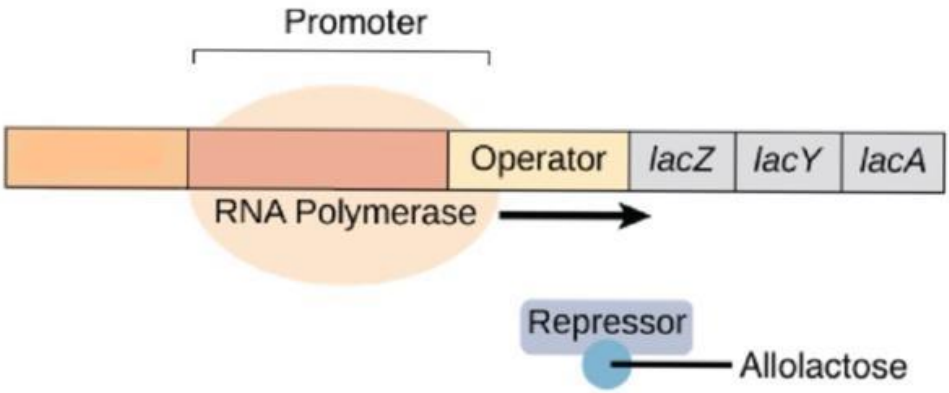
No lactose:

When lactose is absent, the *lac* repressor binds tightly to the operator. It gets in RNA polymerase's way, preventing transcription.



With lactose:

Allolactose (rearranged lactose) binds to the *lac* repressor and makes it let go of the operator. RNA polymerase can now transcribe the operon.



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Lac Operon : Gene Control in Prokaryotes

* An operon is a length of DNA which contains a cluster of genes and serves as a unit of gene expression.
* A lac operon controls the expression of genes which code for proteins involved in lactose metabolism in the bacterium E. coli.
* It consists of :

A) A Promoter site to which the RNA Polymerase binds.

B) An Operator site to which the repressor protein binds.

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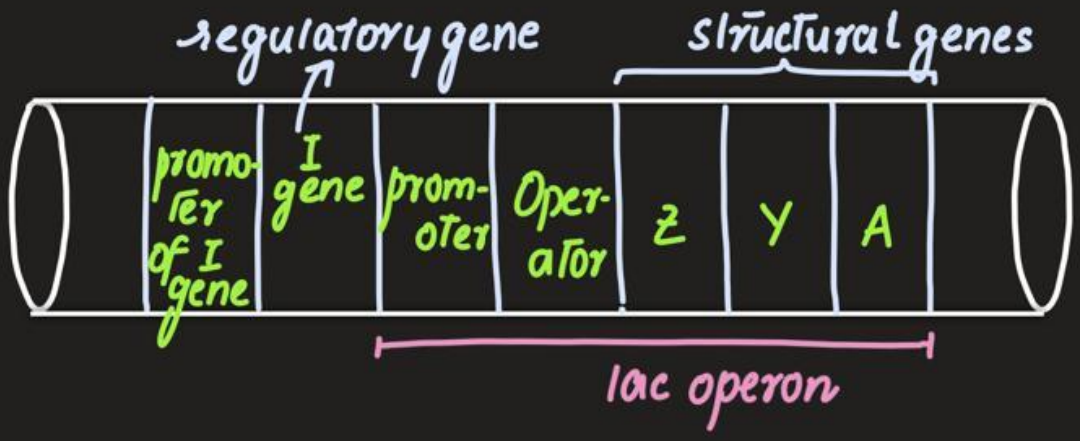
c) Three Structural Genes which include Lac Z, Lac Y, Lac A.

i) Lac Z codes for the enzyme β-galactosidase.

ii) Lac Y codes for the protein Lactose Permease.

iii) Lac A codes for the protein Transacetylase.



* The regulatory gene (I gene) lies close to the promoter region of the operon and codes for a protein known as the repressor protein.

* Repressor protein is an allosteric protein which contains two binding sites:

i) for attachment to the operator region of the operon.

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ii) for attachment to lactose.

* The lac operon is switched on in the presence of lactose and switched off when lactose is absent.

* This ensures that the bacterium E. coli does not synthesise the proteins when they are not required.

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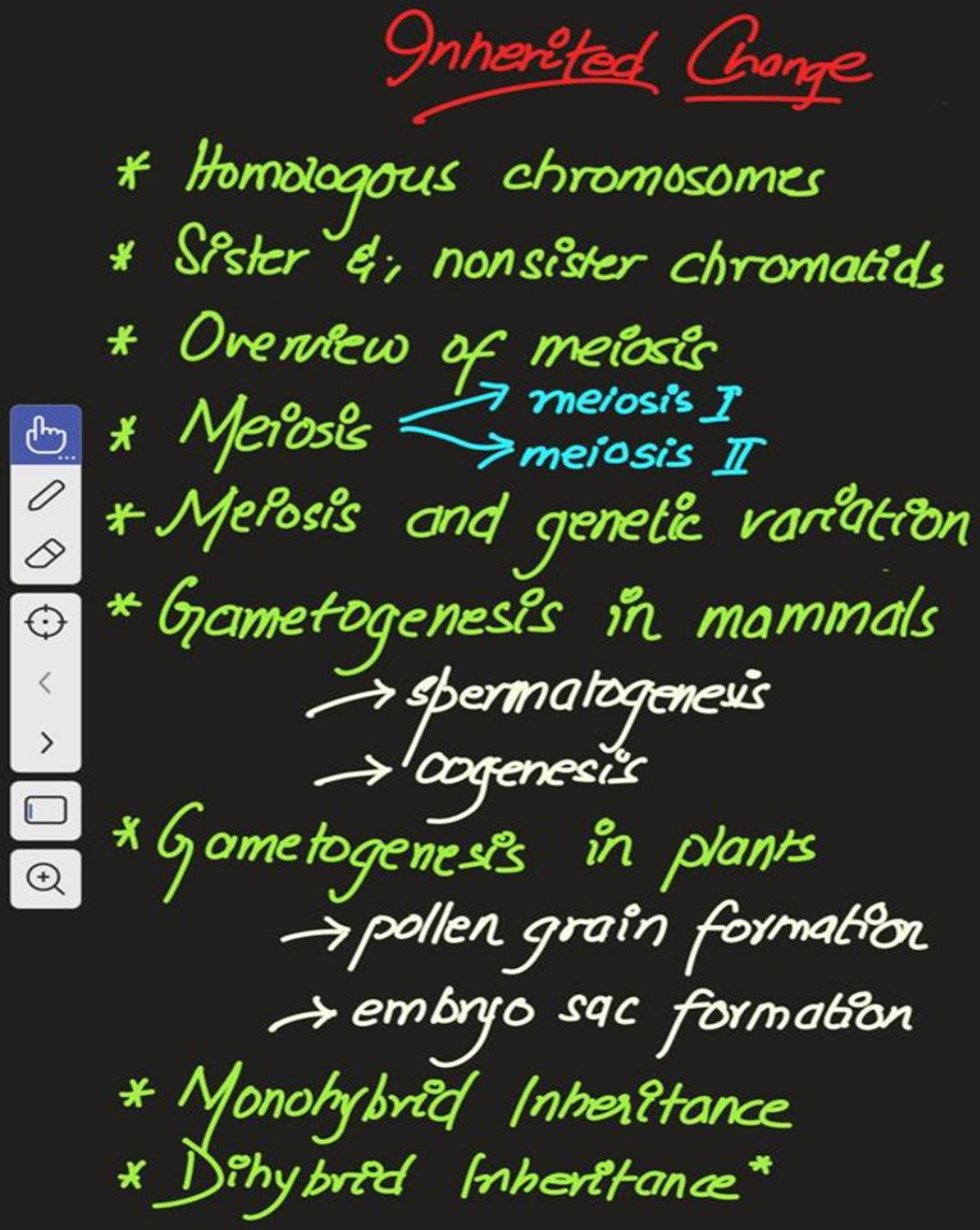

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Gene Control in prokaryotes
⇒ lac operon

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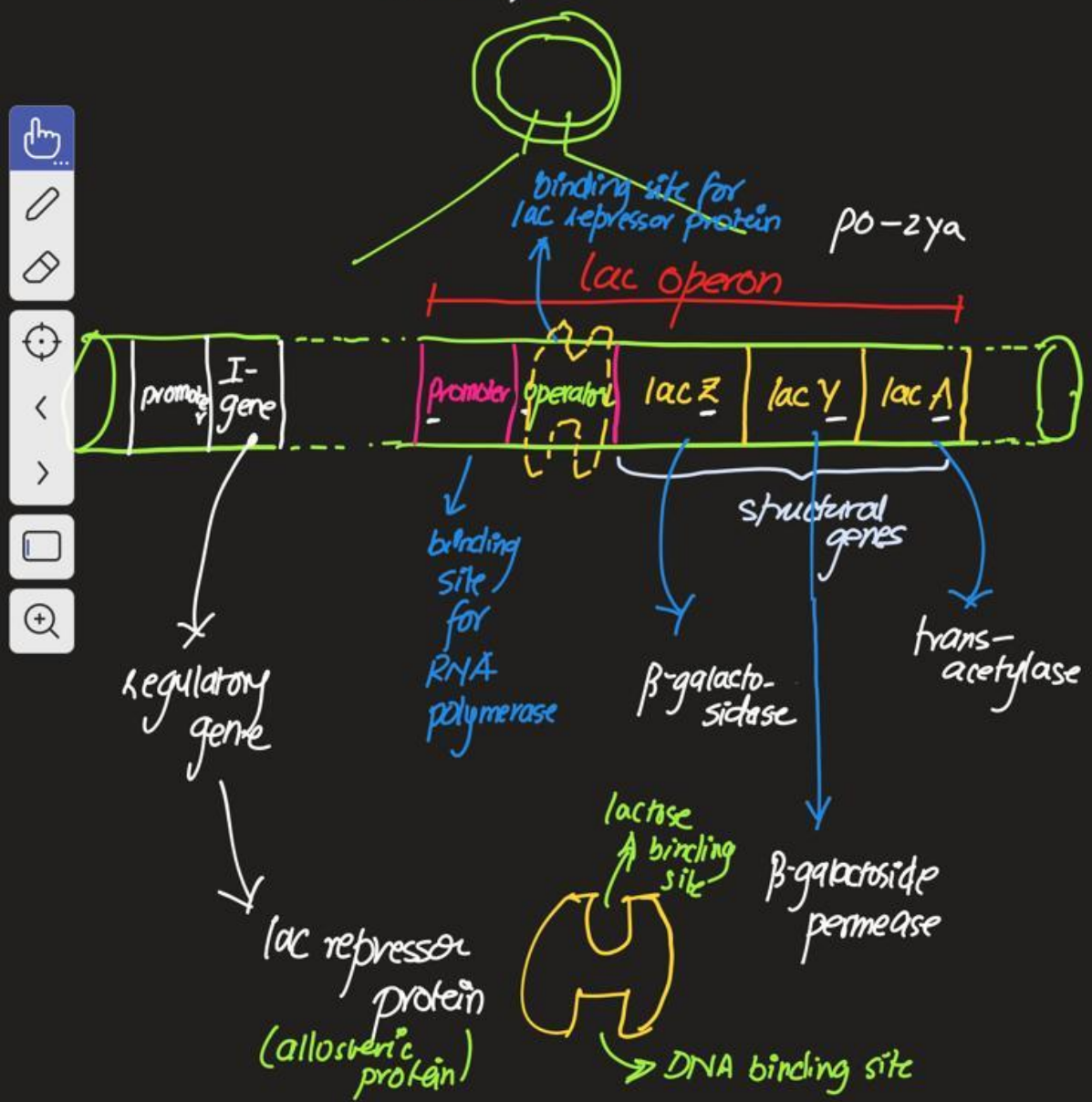
Gene Control in Prokaryotes

⇒ Lac Operon (in E. coli)

(lactose)



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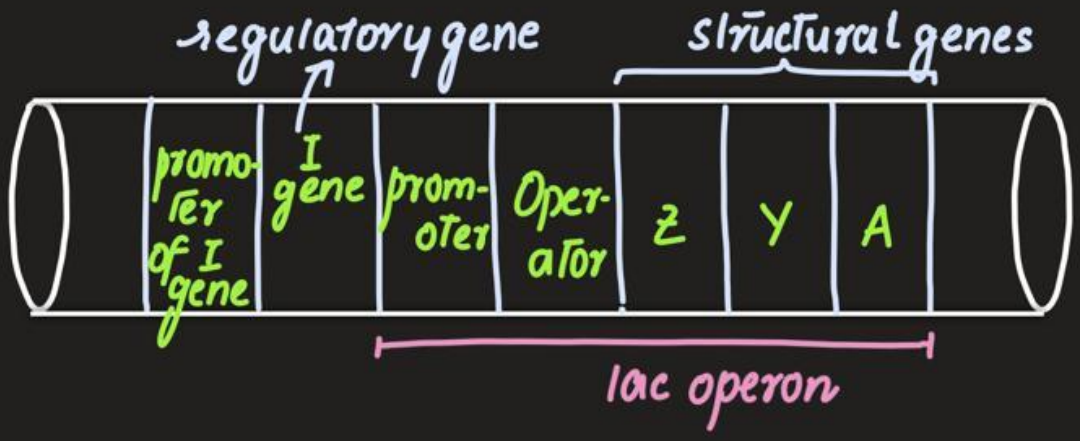

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* Repressor protein is an allosteric protein which contains two binding sites:

i) for attachment to the operator region of the operon.

ii) for attachment to lactose.

* The lac operon is switched on in the presence of lactose and switched off when lactose is absent.

* This ensures that the bacterium E. coli does not synthesise the proteins needed for lactose metabolism when they are not required.

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* When lactose is absent:

① The lac repressor protein binds to the operator region.

② The promoter is blocked and RNA polymerase can't bind to the promoter.

③ Structural genes are therefore NOT transcribed.

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* When lactose is present:

① Lactose binds to the repressor protein causing a change in its shape (allosteric effect).

② This change in shape unbinds the repressor from the operator thereby unblocking the promoter.

③ RNA polymerase can now bind to the promoter and transcribe the structural genes Lac Z, Lac Y and Lac A.

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ⓐ Lac Z codes for β-galactosidase which breaks down lactōse into glucose & galactōse.

ⓑ Lac Y codes for Lactōse Permease which is a transmembrane protein tō allow uptake of lactōse through the phospholipid bilayer of E. coli.

ⓒ Lac A codes for Transacetylase, the function of which is NOT clearly known.

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I-gene & constitutive proteins

* I-gene is a regulatory gene which is expressed at all times in a cell. Such genes are said to have a constitutive expression.
* The protein products of such genes are therefore termed as constitutive proteins.
* These proteins are produced all the time because the cell needs them at all times.
* Lac repressor protein is a constitutive protein.

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Inducible & Repressible enzymes

* β-galactosidase is an example of inducible enzyme. An enzyme is said to be inducible if its production is 'switched on' in the presence of its inducer (usually its substrate). In case of β-galactosidase, lactose is the inducer.

* Unlike β-galactosidase, some enzymes are repressible. These enzymes are produced by the cell by default but their production can be stopped in the presence

of a repressor protein.

* The benefit of gene control in prokaryotes (using a lac operon for example) is that the bacteria does NOT waste amino acids or ATP in synthesizing proteins NOT needed by the cell.

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Gene Control in Eukaryotes

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Gene Control in Eukaryotes

1. Only 2% of the human DNA consists of gene coding for proteins with functional role. These are **structural genes**.

2. Of the remaining 98%, some of the DNA consists of **regulatory genes** and **control sequences** (such as promoters) that control gene expression.

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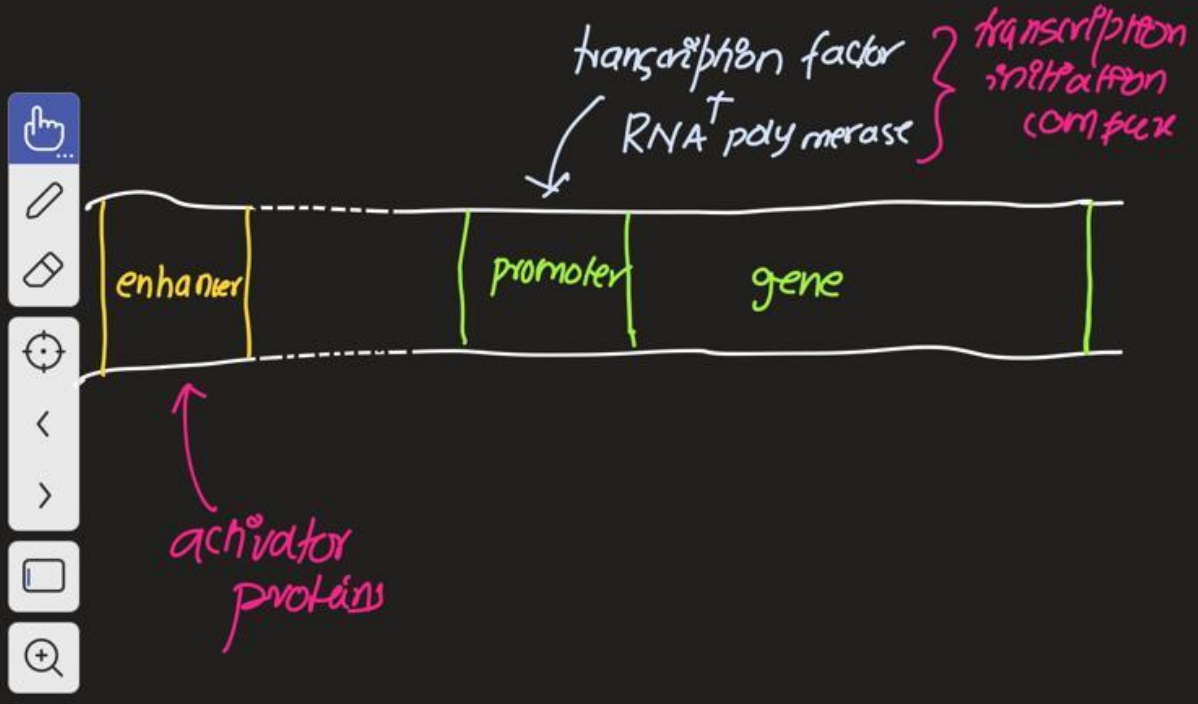
Gene Expression in Eukaryotes



enhancer + promoter → eg's. of control sequences

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Q : How does a gene express (transcribe) itself in an eukaryote ?

Ans : * Transcription is controlled by general transcription factors in eukaryotes.

* These general transcription factors bind to the promoter region located before the start of the gene.

* The enzyme RNA polymerase thereafter binds to the promoter to form the Transcription initiation complex (transcription factors + RNA polymerase bound to promoter).

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* Transcription of the template strand of the gene begins once the transcription complex has assembled.

* The rate of transcription can be increased when the activator proteins bind to enhancer site on the DNA (located upstream).

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* enhancer: binding site for activator proteins
* promoter: binding site for:
  1. general transcription factors
  2. RNA polymerase

  \+ } transcription initiation complex

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Q : Which genes code for transcription factors?

Ans: Regulatory genes

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Cell cycle & growth of organisms is controlled by Transc. factors ②

Sex determination is achieved via transcription factor ①

Gene control in eukaryotes via transcription factors → examples

③ Response to external stimuli is also controlled by Transc. factors

④ Gibberellins & their role in seed germination

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Transcription factors in eukaryotes → EXAMPLES

Some examples of the significance of transcription factors in eukaryotes are as follows:

- Testicular determining factor (TDF) is a transcription factor coded by the TDF gene on the Y chromosome. This transcription factor is responsible for the determination of male sex in humans.

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② Certain transcription factors coded by tumor suppressor genes and/or protooncogenes control cell cycle and growth in eukaryotic organisms.

③ Transcription factors enable eukaryotes to respond to external environmental stimuli by 'switching on' or 'switching off' genes.

④ Certain plant hormones such as gibberellins exert their effect via transcription factors.

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Seed germination & role of gibberellins

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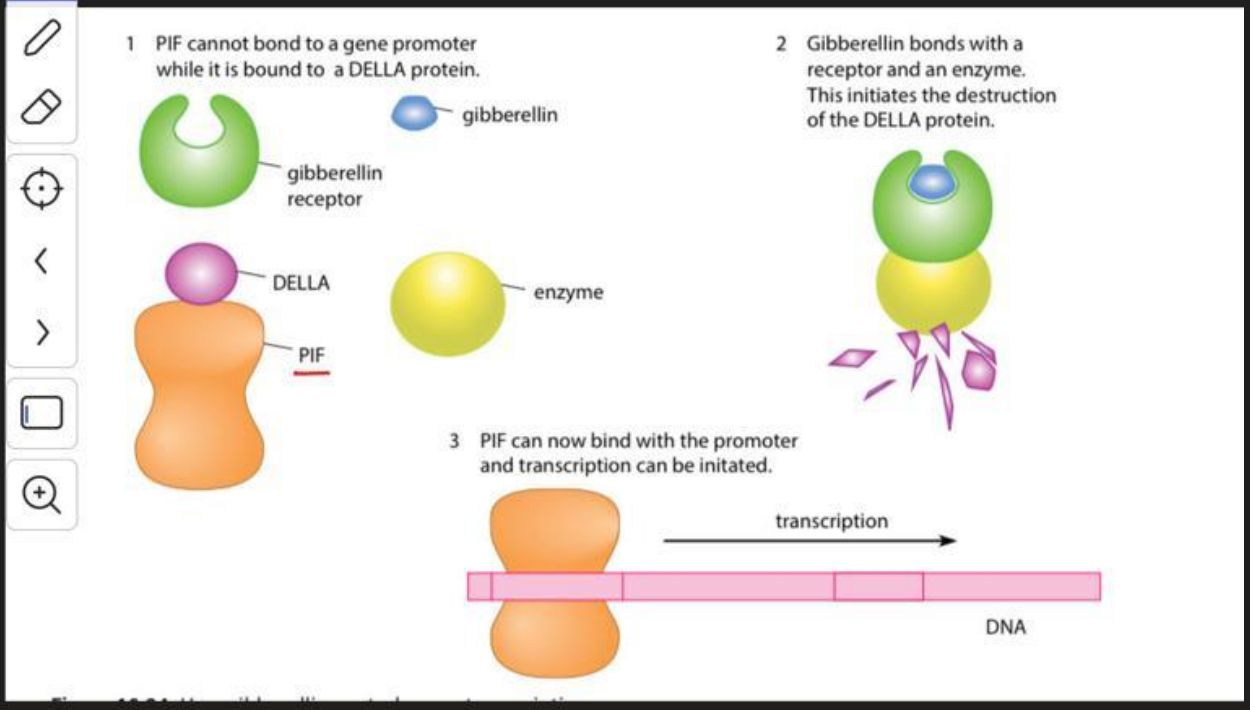
Seed germination & role of gibberellins



PIF → phytochrome interacting factor

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SEED GERMINATION & ROLE OF GIBBERELLINS

1- Gibberellins are responsible for promoting seed germination by enabling the Transcr. iption of the gene which codes for the α- amylase enzyme.

2. Gibberellins carry out this effect by promoting the destruction of DELLA proteins which inhibits a transcription factor from binding to the promoter of the α- amylase gene.

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3. The transcription factor is the phytochrome interacting factor (PIF) which is inhibited when bound to DELLA proteins.

- Gibberellins bind to their receptors and activate an enzyme which causes the breakdown of DELLA proteins, thereby enabling the PIF to promote the transcription of α- amylase gene by binding to the gene promoter.

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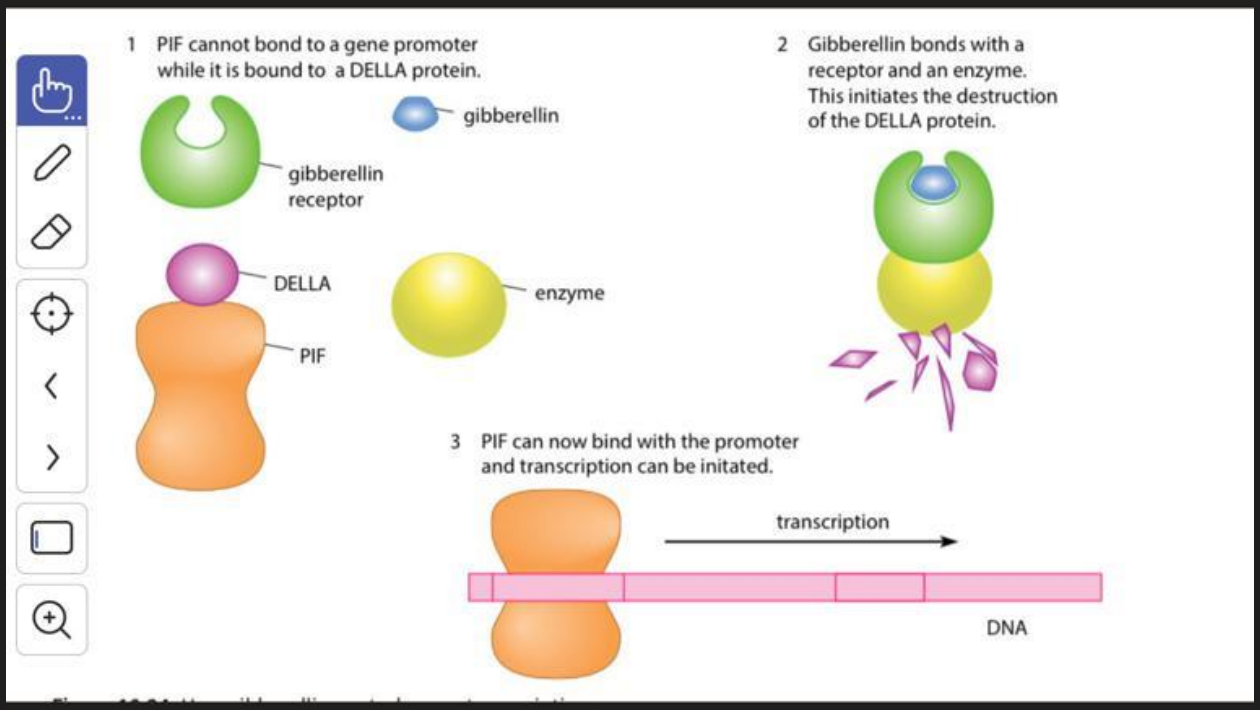
1 PIF cannot bond to a gene promoter while it is bound to a DELLA protein.
gibberellin
gibberellin receptor
DELLA
PIF
enzyme
2 Gibberellin bonds with a receptor and an enzyme. This initiates the destruction of the DELLA protein.
3 PIF can now bind with the promoter and transcription can be initated.
transcription
DNA

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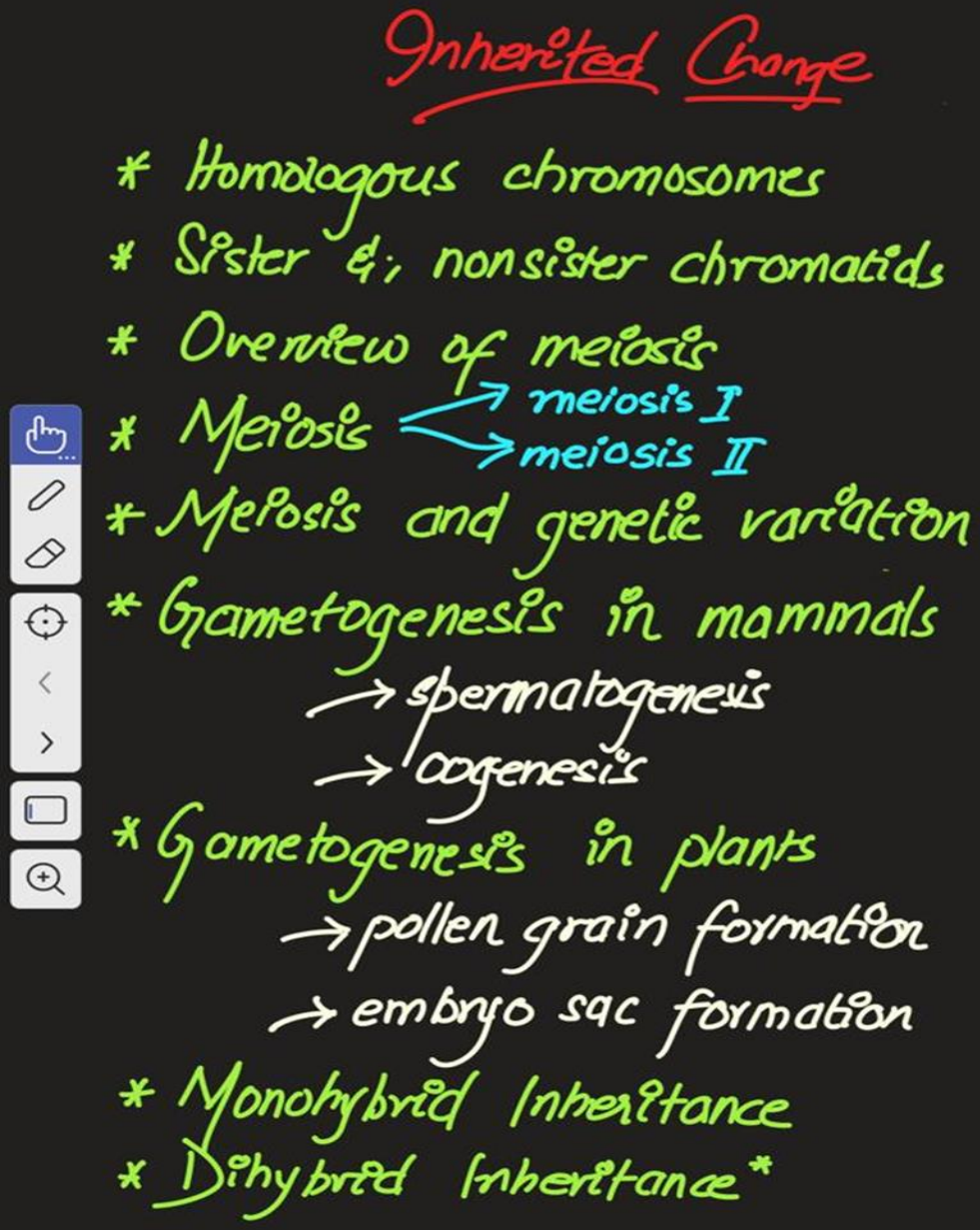

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Mutations & Changes in phenotype

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Mutations and changes in Phenotype

① Mutations are classified into → Gene Mutation
→ Chromosomal Mutations

② We discussed gene mutations in detail in AS.

③ Chromosomal mutation refers to a change in the structure or whole number of chromosomes. Examples of diseases which result due to a chromosomal mutation are: ⓐ Down's Syndrome (47 chromosomes - extra chromosome 21)
ⓑ Turner's Syndrome (45 chromosomes - one less X chromosome in females)

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Q: Describe the ways by which gene mutation can occur? (6)

Ans: A gene mutation is defined as an alteration in the sequence of nucleotides of a particular gene which may alter the primary structure of the polypeptide. There are three different types of gene mutations, namely: 1) Base substitution which involves replacement of a normal nucleotide with a mutant nucleotide having a different base

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2) Base addition which involves addition of one or more nucleotides in a particular gene.

3) Base deletion which involves removal of one or more nucleotides from a particular gene.

Base addition and base deletion may lead to frameshift mutation. These mutations occur during DNA replication which leads to formation of mulānt allele. At times a gene mutation may result due to a

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spontaneous (random) event such as exposure To ionising or ultraviolet radiation.

* We will be discussing three diseases as examples of gene mutations;

1) Sickle cell Anaemia (autosomal recessive)

2) Albinism (autosomal recessive)

3) Huntington's disease (autosomal dominant)

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Sickle Cell Disease

* Is an autosomal recessive disorder.
* Results due to a gene mutation which involves base substitution.
* Mutation occurs in the gene that codes for the β-globin polypeptide of the protein Haemoglobin.
* The sixth amino acid in the normal β-globin chain is glutamic acid coded by the triplet CTC. A base substitution changes CTC to CAC which codes for

amino acid Valine.

* Glutamic acid is hydrophilic while valine is hydrophobic. This change in nature of amino acid causes haemoglobin to become less soluble and precipitate when not bound to oxygen. These haemoglobin molecules stick with each other to form long fibres which exert a pull on the RBC membrane causing them to become Sickle-shaped.

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* The sickle-shaped RBCs have reduced oxygen carrying capacity thereby causing symplōms of Anaemia.

* Signs and symplōms include fatigue, shortness of breath and myocardial infarctions in severe cases.

* Allele which causes sickle cell anaemia is Hb^S whereas the normal allele is Hb^A.

* Individuals with sickle cell anaemia have the genolypē Hb^SHb^S.

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* Individuals with Hb^AHb^S genotype have sickle cell trait. These individuals have some sickle-shaped RBCs in their blood but don't develop signs and symptoms of sickle cell anaemia.
* Individuals with genotype Hb^AHb^A have no sickle-shaped RBCs in their blood.
* The sickle cell allele is now common in parts of the world where malaria is endemic because individuals with sickle cell trait are less likely to die due to complications of malaria.

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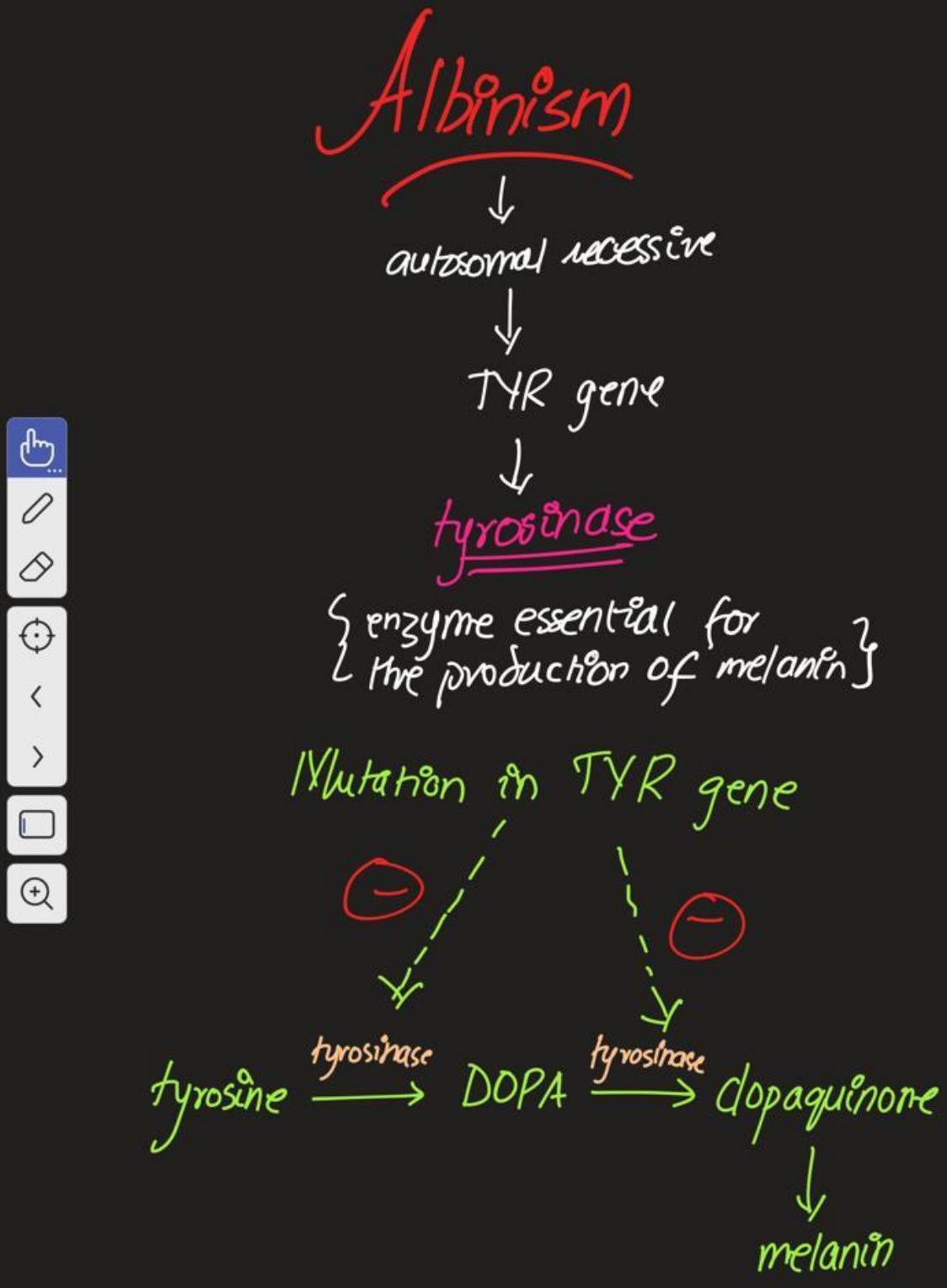
Albinism
autosomal recessive
TYR gene
tyrosinase
{enzyme essential for the production of melanin}
Mutation in TYR gene
tyrosinase
tyrosinase
tyrosine
DOPA
dopaquinone
melanin

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ALBINISM

* May result due to many different mutations, most common of which is an autosomal recessive one.
* The mutations occur in the gene (TYR gene) which codes for enzyme Tyrosinase.
* Tyrosinase is essential for production of melanin in melanocytes.
* Tyrosinase is a transmembrane protein present in the membrane of melanosomes within the melanocytes.

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* Tyrosinase is involved in the production of melanin, through the following bio-chemical pathway:

Tyrosine —(Tyrosinase / tyrōsinase)→ DOPA —(Tyrosinase)→ dopaquinone

↓

melanin

* The gene mutation which causes albinism leads to the production of inactive Tyrosinase or no Tyrosinase at all, which thereafter reduces or stops melanin production.

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* The signs and symptoms of albinism include:

A. Pale skin colour and pale hair colour
B. Pink or light blue iris
C. Rapid movement of the eyes (nystagmus)
D. Poor vision
E. Photophobia
F. Skin burns & increased susceptibility to skin cancers.

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Q: Explain how the presence of a mulānt allele results in albinism?

Ans: Albinism is an autosomal recessive disease. It only occurs in individuals homozygous for the mutant recessive allele of the gene TYR.

The dominant allele of the TYR gene codes for a functional copy of the enzyme tyrosinase. This enzyme is located in the membrane of organelles known as melanosomes. These organelles are found in melanin producing cells known as

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melanocytes. Tyrosinase catalyses the reactions in the biochemical pathway involved in production of melanin:

Tyrosine —(Tyrosinase / tyrosinase)→ DOPA —(Tyrosinase)→ dopaquinone → melanin

The mutant recessive allele of TYR gene codes for a non-functional tyrosinase which leads to reduced or no melanin production. The phenotypic changes associated with the disorder include fair skin and fair hair colour.

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Huntington's Disease (→ chorea)

↓

autosomal dominant

↓

MOST with HD have a heterozygous genotype

Dd x dd

D d d

Dd dd

1 : 1

↓

gene mutation in the HTT gene → codes for a protein huntingtin

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HUNTINGTON'S DISEASE

* It is an autosomal dominant disorder.
* Occurs due to a triplet repeat mutation in the gene HTT that codes for the protein Huntingtin. This gene is present on chromosome number 4.
* Most individuals with Huntington's disease are heterozygous therefore there's a 50% likelihood that a child born will also have Huntington's disease.

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* The triplet repeat mutation involves a large number of CAG repeats in an unstable segment of the gene which codes for the protein Huntingtin. These repeats are also termed as stutters.

* There's a roughly inverse correlation b/w the no. of repeats and the age of onset of Huntington's disease. The greater the number of repeats, the earlier the age of onset.

* Symptoms usually manifest at middle age.

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* Signs and symptoms include:
  A. Loss of brain cells
  B. Enlargement of brain ventricles
  C. Involuntary muscular movements termed as chorea.
  D. Dementia (memory loss)

* Huntingtin is essential for the normal functioning of neurones.