

ULTIMATE KCET

CRASH COURSE 2026

Botany

Lecture - 01

Principles of Inheritance and Variation

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Topics *to be covered*

1 SYNOPSIS

2 QUESTIONS

3

4





Important topics

7 years 1856-1863
Pisum sativum - short life cycle, self pollinated
 many seeds, easily grown

7

Character	Dominant trait	Recessive trait
Seed shape	Round	Wrinkled
Seed colour	Yellow	Green
Flower colour	Violet	White
Pod shape	Full	Constricted
Pod colour	Green	Yellow
Flower position	Axial	Terminal
Stem height	Tall	Dwarf

S.No.	Characters	Contrasting Traits
1.	Stem height	Tall/dwarf π (14)
2.	Flower colour	Violet/white π
3.	Flower position	Axial/terminal π
4.	Pod shape	Inflated/constricted
5.	Pod colour	Green/yellow
6.	Seed shape	Round/wrinkled
7.	Seed colour	Yellow/green

- Laws of independent assortment
- Law of segregation
- Law of dominance

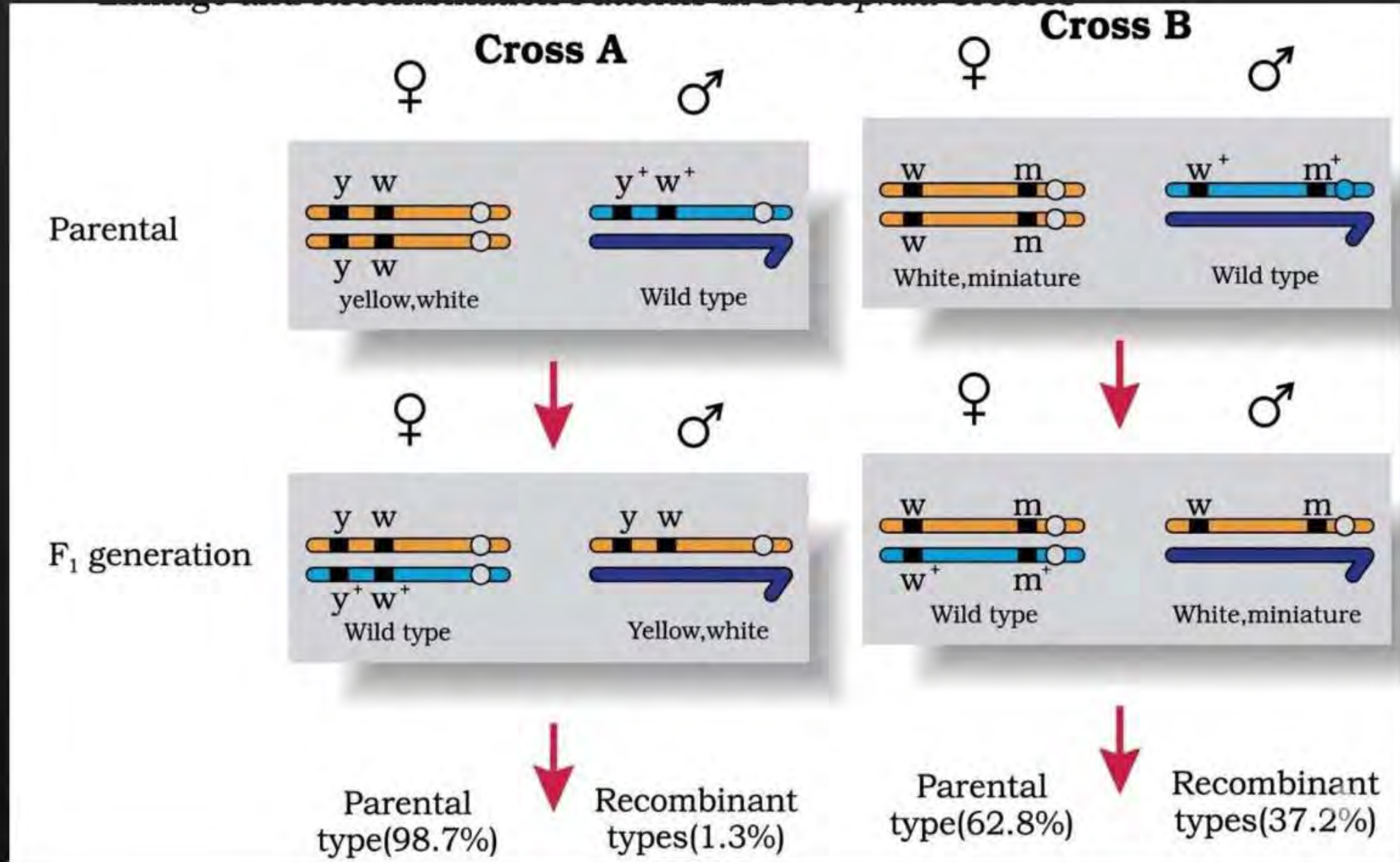
- Ratio. monohybrid cross. 3:1
- dihybrid cross 9:3:3:1
- Test cross 1:1



Important topics

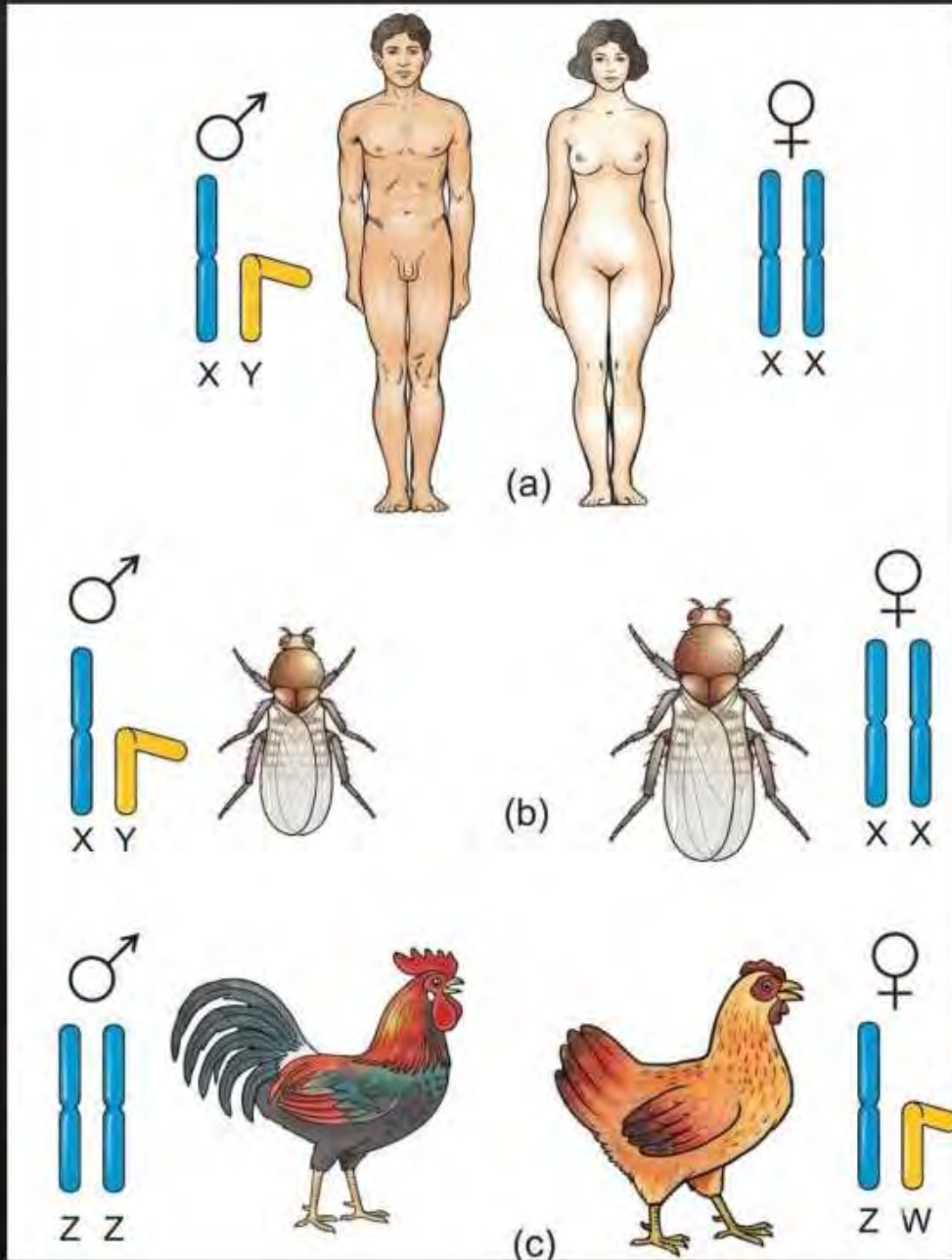
- Multiple allelism - ABO blood grouping
- Pleiotropy - PKU, SCA. Starch synthesis gene (B gene)
- Co-dominance - AB blood group
- Polygenic inheritance. Skin colour in humans
- Incomplete dominance Flower Colour in Snapdragon 1:2:1

Important topics







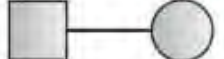

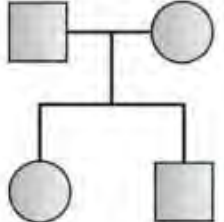
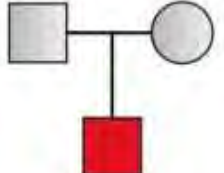



Important topics





Important topics

	Male
	female
	sex unspecified
	affected individuals
	mating
	mating between relatives (consanguineous mating)
	parents above and children below (in order of birth-left to right)
	parents with male child affected with disease
	five unaffected offspring



Important topics

- Mendelian Disorders - Most common and prevalent Mendelian disorders are Hemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalassemia.
- Chromosomal Disorders - Down's syndrome Turner's syndrome, Klinefelter's syndrome. Symptoms Polyploidy, aneuploidy

Question



Match Column - I with Column - II and find the correct answer:

Column - I		Column - II	
1.	Aneuploidy	p.	Increase in whole set of chromosome
2.	Monoploidy	q.	Loss or gain of a chromosome
3.	Polyploidy	r.	Two sets of chromosomes
4.	Diploidy	s.	A single set of chromosomes

A 1-p 2-q 3-r 4-s

B 1-r 2-p 3-q 4-s

C 1-q 2-s 3-p 4-r

D 1-s 2-r 3-p 4-q

Question



A human male is heterozygous for autosomal genes 'A' and 'B'. He is also hemizygous for hemophilic gene 'h'. What percentage of sperms will carry 'abh' genotype?

- A** 25%
- B** 50%
- C** 75%
- D** 0%

Question



The gene for haemophilia is located on 'X' chromosome. Hence it is normally impossible for a

- A** haemophilic father to pass the gene to his daughter.
- B** carrier mother to pass the gene to her daughter.
- C** carrier mother to pass the gene to her son.
- D** haemophilic father to pass the gene to his son

Question



Example for autosomal hyper aneuploidy is:

- A** Down's syndrome
- B** Klinefelter's syndrome
- C** Turner's syndrome
- D** Haemophilia

Question



In dihybrid cross, when F_1 plants ($RrYy$) are self hybridized, the ratio of segregation of yellow and green in F_2 is:

- A** 1:2:1
- B** 3:1 ✓
- C** 9:3:3:1
- D** 1:1:1:1

Question



Replacement of which one of the following nucleotides in the gene causes sickle cell anaemia ?

- A** A to T
- B** T to A
- C** U to A
- D** C to G

Question



The gene disorder phenylketonuria is an example for

- A** Polygenic inheritance
- B** Pleiotropy
- C** Multiple allelism
- D** Multiple factor

Question



Identify the odd one among the following disorders:

- A** Sickle-cell Anaemia
- B** Thalassemia
- C** Haemophilia
- D** Phenyl Ketonuria

Question



From the Chromosomal Complements given below, identify the one which shows female heterogamety.

- A** $XX-XY$
- B** $ZZ-ZW$
- C** $XX-XO$
- D** $XX-XXY$

Question

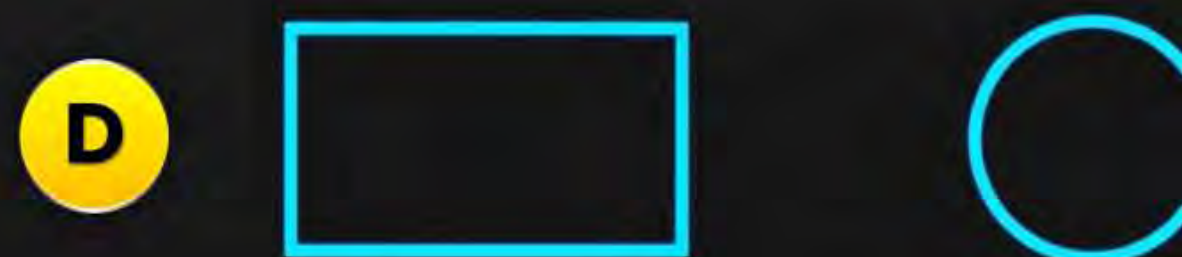
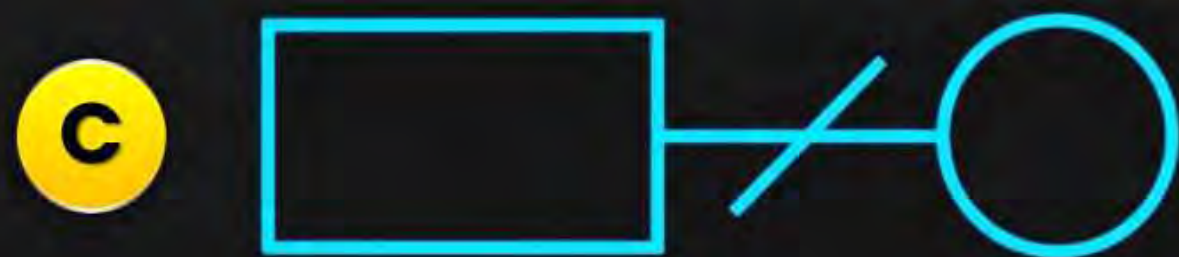
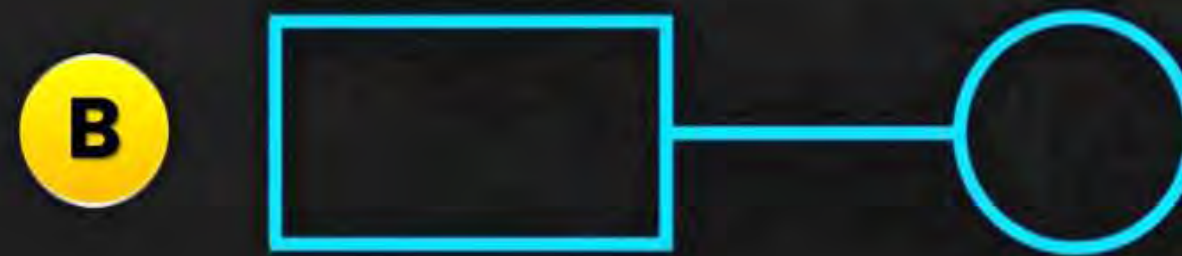


In Morgan's experiment with *Drosophila*, when yellow bodied white eyed female was crossed with brown bodied red eyed male and their F_1 progeny were intercrossed. What was the percentage of recombinants in F_2 generation?

- A** 98.7%
- B** 37.2%
- C** 62.8%
- D** 1.3%

Question

In the following symbols, used in human pedigree Analysis, identify the symbol that denotes consanguineous mating.



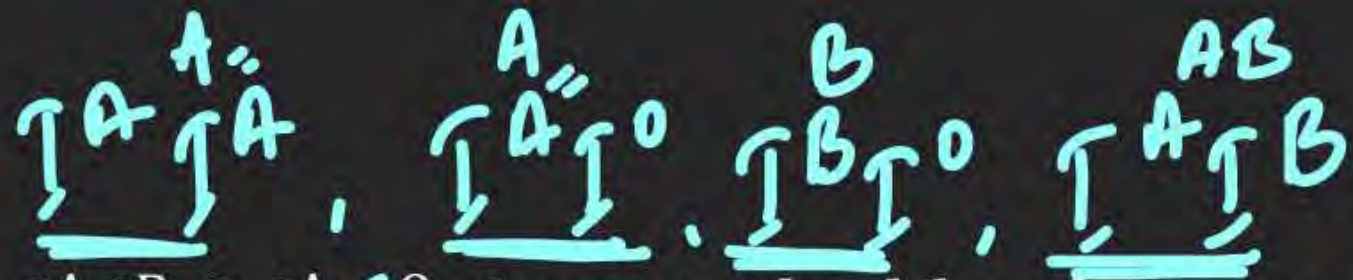
Question



In one of the hybridisation experiments, a homozygous dominant parent and a homozygous recessive parent are crossed for a trait. (Plant shows Mendelian inheritance pattern)

- A** Dominant parent trait appears in F_1 generation and recessive parent trait appears in F_1 and F_2 generations. ✗
- B** Dominant parent trait appears in F_1 generation and recessive parent trait appears in F_2 generation ✓
- C** Dominant parent trait appears in F_2 generation and recessive parent trait appears only in F_1 generation ✗
- D** both F_1 & F_2 generations, ~~recessive~~ Dominant parent trait appears in both F_1 & parent trait appears in only F_2 generation ✓

Question



The genotype of a husband and wife are $I^A I^B$ & $I^A i$. Among the blood types of their children, how many different genotypes & phenotypes are possible?

- A** 3 genotypes; 3 phenotypes
- B** 4 genotypes; 3 phenotypes ✓
- C** 4 phenotypes; 3 genotypes
- D** 4 phenotypes; 4 genotypes

4 pheno

6 genotypes

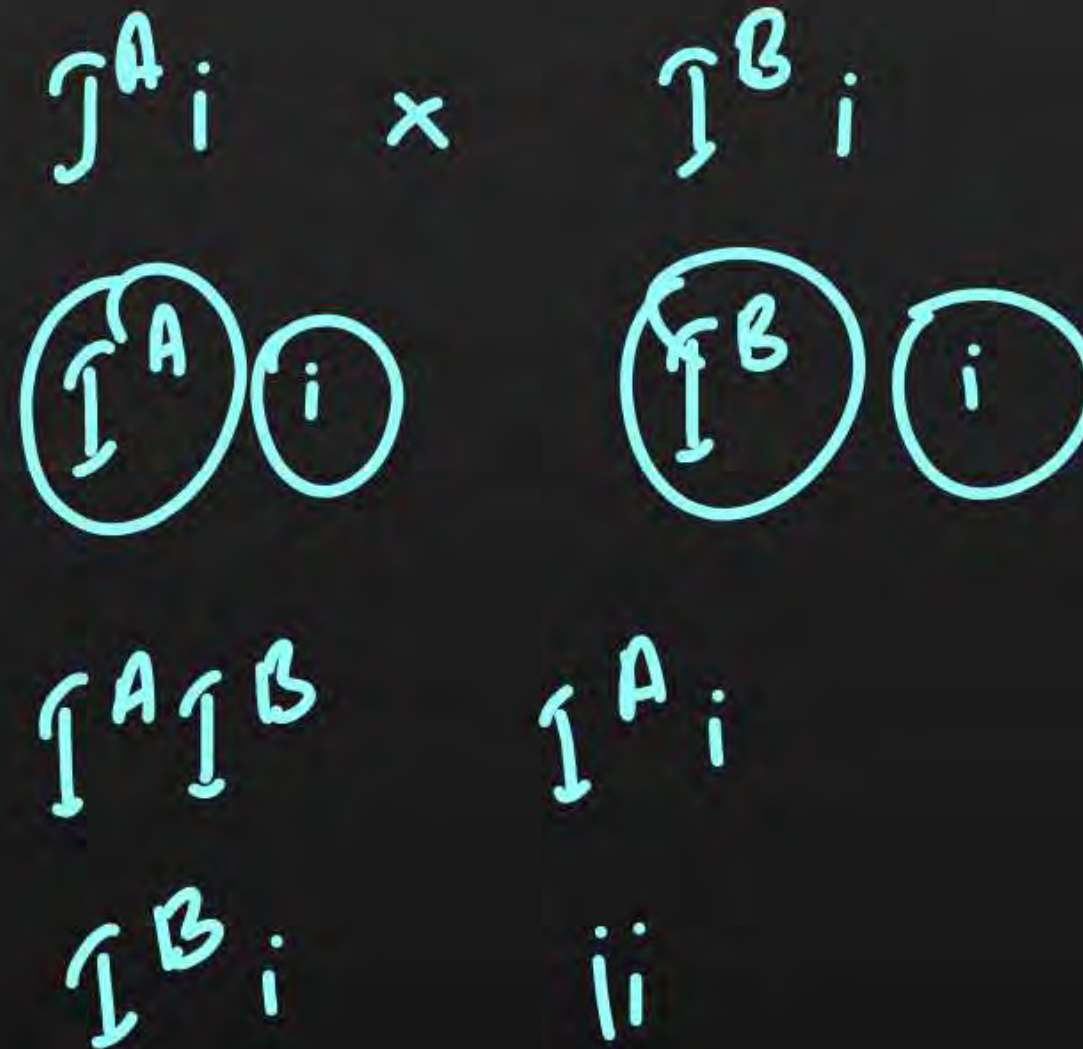
A	$I^A I^A / I^A i$
B	$I^B I^B / I^B i$
AB	$I^A I^B$
O	ii

Question



What is the possible blood group of children whose parents are heterozygous for A & B blood groups?

- A** A, B only
- B** A, B, AB & O ✓
- C** AB only
- D** A, B & AB only



Question



Match the column – I with Column - II

Column - I		Column - II	
i.	Autosomal trisomy	p.	Turner's Syndrome
ii.	Allosomal trisomy	q.	Mendelian disorder
iii.	Allosomal Monosomy	r.	Klinefelter's Syndrome
iv.	Cystic fibrosis	s.	Down's Syndrome

I II III IV I II III IV

A

P q r s

B

P q r s

C

S r q p

D

S r q p

Question



Which among the following characters selected by Mendel in a pea plant is a recessive character?

A Inflated (full) pod - *d*

B Green pod colour - *d*

C White flower - *π*

D Axillary flower - *d*

green seed - 'π'

Question



XO type of sex determination and XY type of sex determination are the examples of

- A** Male Homogamety
- B** Female Homogamety
- C** Male Heterogamety.
- D** Female Heterogamety

Question



Example for Non-Mendelian disorder:

- A** Thalassemia
- B** Haemophilia
- C** Cystic fibrosis
- D** Down's syndrome

Question



Gynecomastia is a symptom of

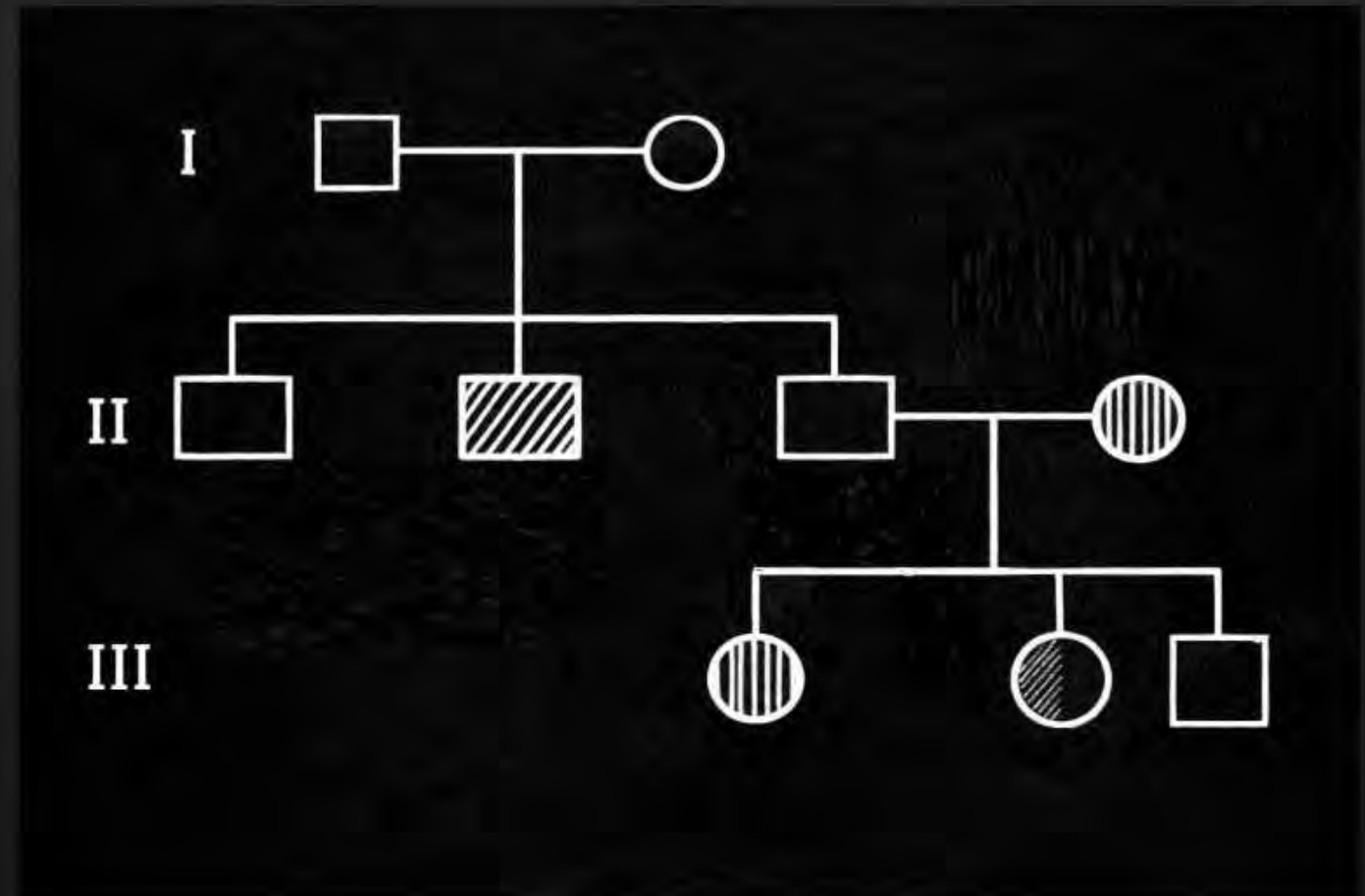
- A** Turner's syndrome
- B** Down's syndrome
- C** Cri-du-chat syndrome
- D** Klinefelter's syndrome

Question



From the following pedigree chart of a family, one can make an analysis that,

- A** It is an autosomal dominant trait
- B** It is an autosomal recessive trait.
- C** It is an allosomal dominant trait.
- D** It is an allosomal recessive trait



Question



In an Organism, mutation in a single gene exhibits multiple phenotypic expressions. Identify the underlying genetic mechanism in the above instance.

- A** Pleiotropy ✓
- B** Incomplete dominance
- C** Polygenic inheritance
- D** Multiple allelism

Question



A pure breeding pea plant with round yellow seeds was crossed with pea plant having wrinkled green seeds. On selfing of F_1 hybrid of this cross, 64 progenies were obtained in F_2 generation. Find out the number of F_2 progenies showing non-parental characters.

A 36

B 4

C 12

D 24

$$F_2 = 64$$
$$3+3=6$$

$$16 - 3$$
$$64 - ?$$

$$\frac{64 \times 3}{16} =$$

Question



A man with blood group A marries a woman having blood group B. The maximum possible blood groups among their progenies are



- A** AB only
- B** A, B, AB
- C** A, B
- D** A, B, AB, O



Question



Which of the following characters was not studied by Mendel in his Pea plant experiments?

- A** Stem height
- B** Pod shape
- C** Seed shape
- D** Leaf shape ✓

Question



Choose the possible genotypes responsible for lightest skin colour in human beings.

A AABBBCC

B AaBbCc

C aabbcc

D AABbCc

Question



Both male and female have normal vision though their fathers were colour blind, and mothers did not have any gene for colour blindness. The probability of their daughter becoming colour blind is

- A** 0%
- B** 15%
- C** 25%
- D** 50%

Question



In male heterogametic type of sex determination

- A** Male parent produces dissimilar gametes.
- B** Male parent produces similar gametes.
- C** Males do not produce gametes.
- D** Female parent produces dissimilar gametes

Question



Which of the following statements is correct?

- A** Sickle cell anaemia is a quantitative problem.
- B** Thalassemia is a qualitative problem.
- C** Female carrier for haemophilia may transmit the disease to sons
- D** Change in whole set of chromosomes is called aneuploidy

Question



Gene-mapping' technology was developed by

- A** Sturtvent
- B** Tschermak
- C** Mendel
- D** Correns

Question



Find the correct statement.

Pleio

- (1) Generally, a gene regulates a trait, but sometimes one gene has effect on multiple traits.
- (2) The trait AB-blood group of man is regulated by one dominant allele and another recessive allele. Hence it is co-dominant. ~~x~~

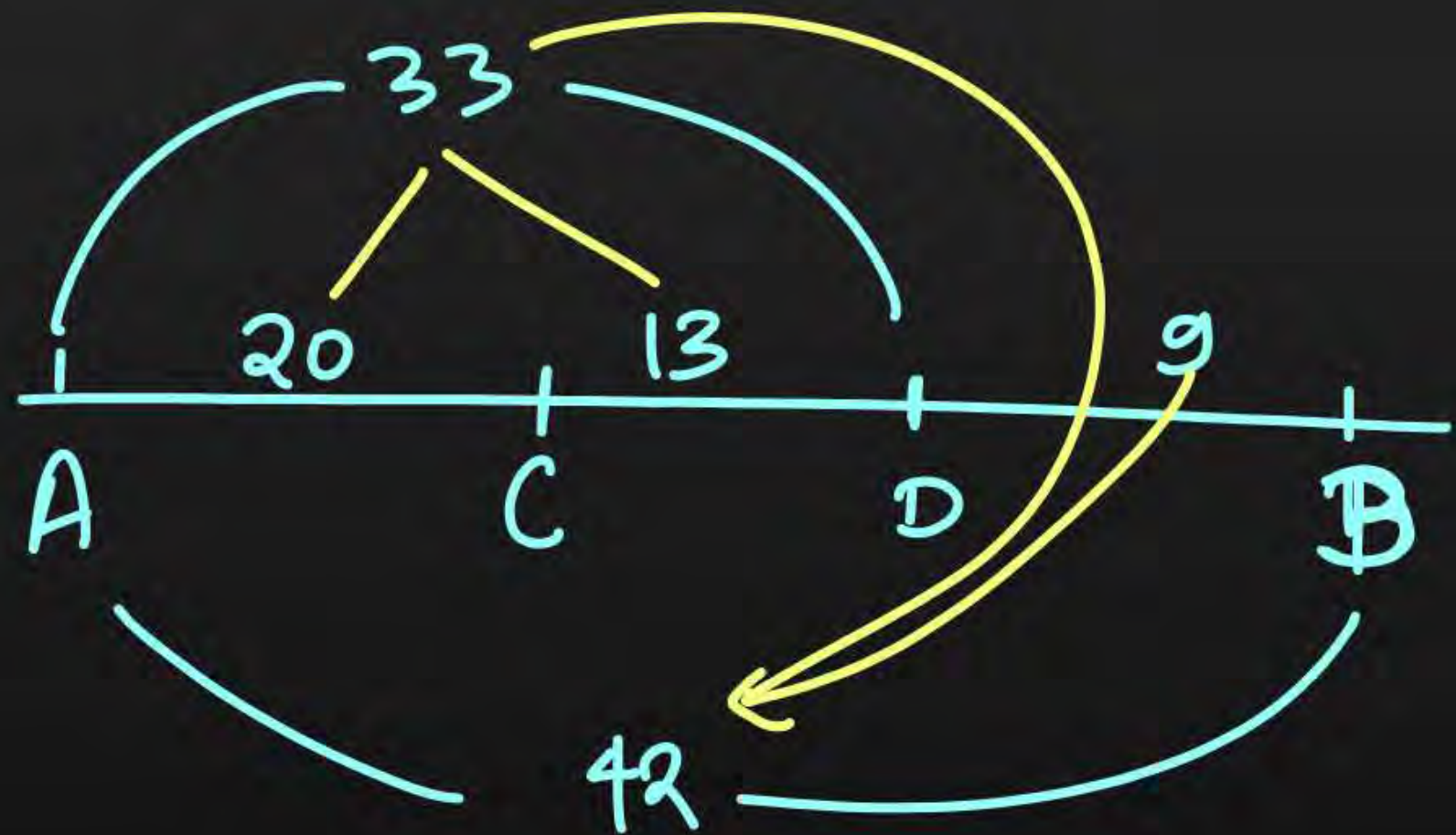
- A** Both Statements (1) and (2) are correct.
- B** Statement (1) is correct. ✓
- C** Both the Statements are wrong.
- D** Statement (2) is correct.

Question



Find the order of genes A, B, C and D on E.coli genophore if the distance between A and D is 33 map units, C and D is 13 map unit A and B is 42 map units, B and D is 9 map unit and A and C is 20 map units

- A** A - B - C - D
- B** A - C - B - D
- C** A - C - D - B
- D** A - D - B - C



Question



In the Mendelian dihybrid cross for the colour and shape of seed, what is the probability of plants having homozygotic round seed character?

A $\frac{3}{4}$

B $\frac{3}{16}$

C $\frac{3}{8}$

D $\frac{1}{4}$

RR

4 - 16

$$\frac{4}{16} =$$

RrYy x RrYy

Rr x Rr = RR : Rr : rr

$\left(\frac{1}{4}\right) : \frac{1}{2} : \frac{1}{4}$

Question



In which of the following cases maximum types of gametes are possible?

- A** AABB
- B** AABb
- C** aaBb
- D** AaBb

$2^n =$
 $n =$ no of heterozygous genes

$$2^2 = 4$$

Question



What is the recombination percentage between gene W (white eye) and m (miniature wing) in *Drosophila* in the experiment conducted by Morgan?

- A** 1.3%
- B** 98.7%
- C** 62.8%
- D** 37.2%



Question



Human male has ___ linkage group more than human female

all linked genes on one chromosome = 1 group

linkage group = haploid chromosomes

humans → ♀ = 23

→ ♂ : $22A + X + Y = 24$

A 24 ✓

B 23 - ♀

C 1

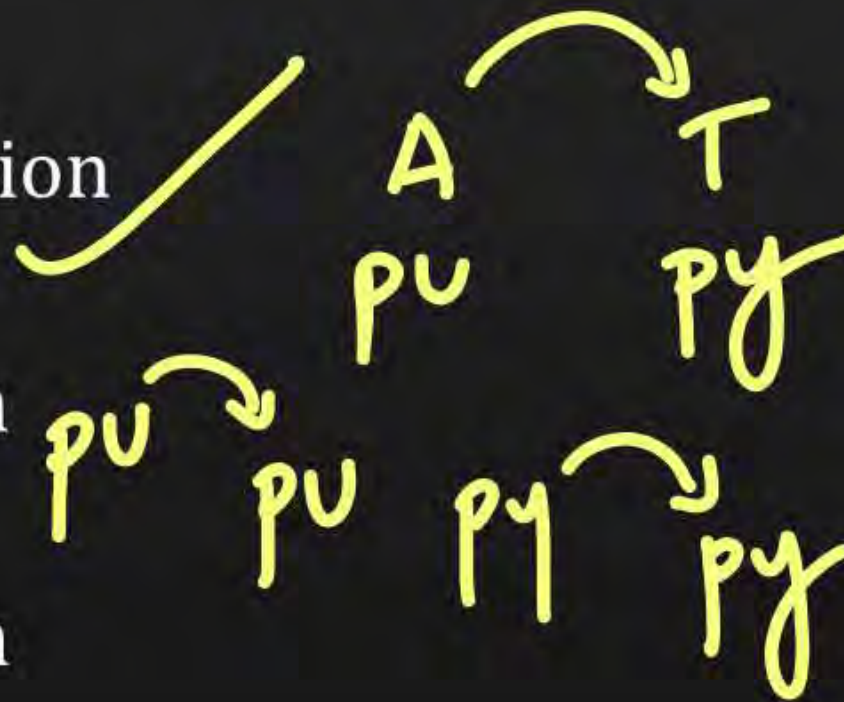
D 0

Question



Sickle cell anaemia is due to

- A** Insertion repeats
- B** Transversion substitution mutation
- C** Transition substitution mutation
- D** Chromosomal shift translocation



Question



Read the following statements and choose correct option

- (A) A gamete carries only one factor of a character ✓
- (B) Starch synthesis in wrinkled seeded pea plants is most efficient ✗
- (C) Modified allele is always the recessive allele ✗

generally

A All are incorrect except A ✓

B All are correct except B

C All are incorrect

D All are correct

Question



Who prepared first genetic map for Drosophila?

- A** Sturtevant ✓
- B** Bateson
- C** Bridge
- D** Muller

Question



Select odd one with respect to Mendelian disorder

- A** Down's syndrome
- B** Haemophilia
- C** Colour blindness
- D** Sickle-cell anaemia

Question



How many types of gametes will be produced from AaBBCcdd ?

A 6

B 8

C 4

D 9

$$2^n = 2^2$$

4



Question



A normal man married a normal woman, both of whom have colour-blind father. What percentage of their daughter would be colour-blind?

- A** 0% ✓
- B** 25%
- C** 50%
- D** 75%



Question



Consider the following cross AABbCcDDEe × AaBbCCDdee Q What will be the proportion of offsprings having geno- type AAbbCCDDEe?

A $\frac{1}{128}$

B $\frac{1}{32}$

C $\frac{1}{64}$

D $\frac{1}{16}$

$$AA \times Aa = AA \quad Aa$$

$\frac{1}{2}$

$$Bb \times Bb = BB ; Bb : bb$$

$\frac{1}{4}$

$$Cc \times Cc = Cc ; Cc$$

$\frac{1}{2}$

$$DD \times Dd = DD ; Dd$$

$\frac{1}{2}$

$$Ee \times ee = Ee ; ee$$

$\frac{1}{2}$

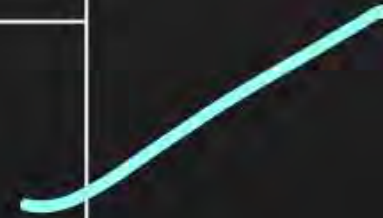
$$\frac{1}{2} \times \frac{1}{4} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2}$$

Question



Find the incorrect match w.r.t pea plant

	Character	Trait	Trait
A	Stem height	Tall <i>d</i>	Dwarf
B	Flower Position	Terminal	Axial
C	Flower colour	Purple <i>d</i>	White
D	Pod shape	Inflated <i>d</i>	Constricted



Question



In F_2 Mendelian population of 100 pea plants, how many are expected to be pure tall?

- A** 100
- B** 75
- C** 50
- D** 25

$$F_2 - 100$$

$$\uparrow T = \frac{1}{4}$$

$$\uparrow t = \frac{1}{2}$$

$$tt = \frac{1}{4}$$

$$\frac{1}{4} \times 100$$

=



Question



From the following postulation of Mendel, which one expresses the 2nd law of Mendel?

- A** Each character is controlled by a discrete unit called factor
- B** Factors occur in pairs
- C** In a dissimilar pair, only dominant factor is expressed
- D** Two factors for a character are not mixed up. Both are recovered in F_2 -population after segregation

↑
↑
↑ tall, dwarf



Question



Which of the following is/are possible for a modified allele?

(i) Can produce no enzyme ✓

(ii) Can produce a non functional enzyme ✓

(iii) Can produce an altered and functional product ✗

A Only (i)

B Only (i) & (ii)

C Only (ii)

D All (i), (ii) & (iii)

Question



A purple flowered ($CcPp$) and a white flowered ($CCpp$) sweet pea plants (*Lathyrus odoratus*) are crossed. Find the ratio of purple {white} flowered plants formed in F₁ generation

$$Pp \times pp$$

$$1:1$$

A 3 : 1

B 1 : 1 ✓

C 2 : 1

D 1 : 3

Question



Consider the following cross $AaBb \times AaBb$. What are the chance of occurrence of a progeny with $Aabb$ genotype

$$Aa \times Aa = AA; Aa; aa$$
$$Bb \times Bb = bb \quad \frac{1}{4} \quad \frac{1}{2}$$

- A** $\frac{1}{4}$
- B** $\frac{1}{8}$
- C** $\frac{1}{16}$
- D** $\frac{1}{32}$

Question



Which of the following related to genetics is universally applicable

- A** Law of dominance
- B** Law of Segregation ✓
- C** Law of independent assortment
- D** Law of incomplete dominance

Question



In ZW-ZZ type of sex determination

- A** Female is homogametic
- B** Male is heterogametic
- C** Female is heterogametic ✓
- D** Male is either homogametic or heterogametic

Question



Sickle cell anaemia is an example of

- A** Polygenic trait
- B** Pleiotropic trait ✓
- C** Qualitative trait ✓
- D** both (2) and (3) ✓

Question



Which characters were not chosen by Mendel in his pea plant experiment?

- A** Height of plant and flower position
- B** Seed shape and seed colour
- C** Flower shape and cotyledon colour
- D** Pod shape and pod colour

Question



If two linked genes are 50 map unit apart, what does it reveal w.r.t. frequency of gene recombination?

- A** It is equal to 50% ✓
- B** It is less than 50%
- C** It is above 50%
- D** Both (1) and (3)

Question



Which type of gametic fusion would give rise to Klinefelter's syndrome?

- A** $(A+XX) \text{ ♀} \times (A+Y) \text{ ♂}$ ✓
- B** $(A+X) \text{ ♀} \times (A+Y) \text{ ♂}$
- C** $(A+XY) \text{ ♀} \times (A+X) \text{ ♂}$
- D** $(A+X) \text{ ♀} \times (A+XX) \text{ ♂}$

Question



Find out the types of gametes that can be formed from the genotype AaBBCcDD?

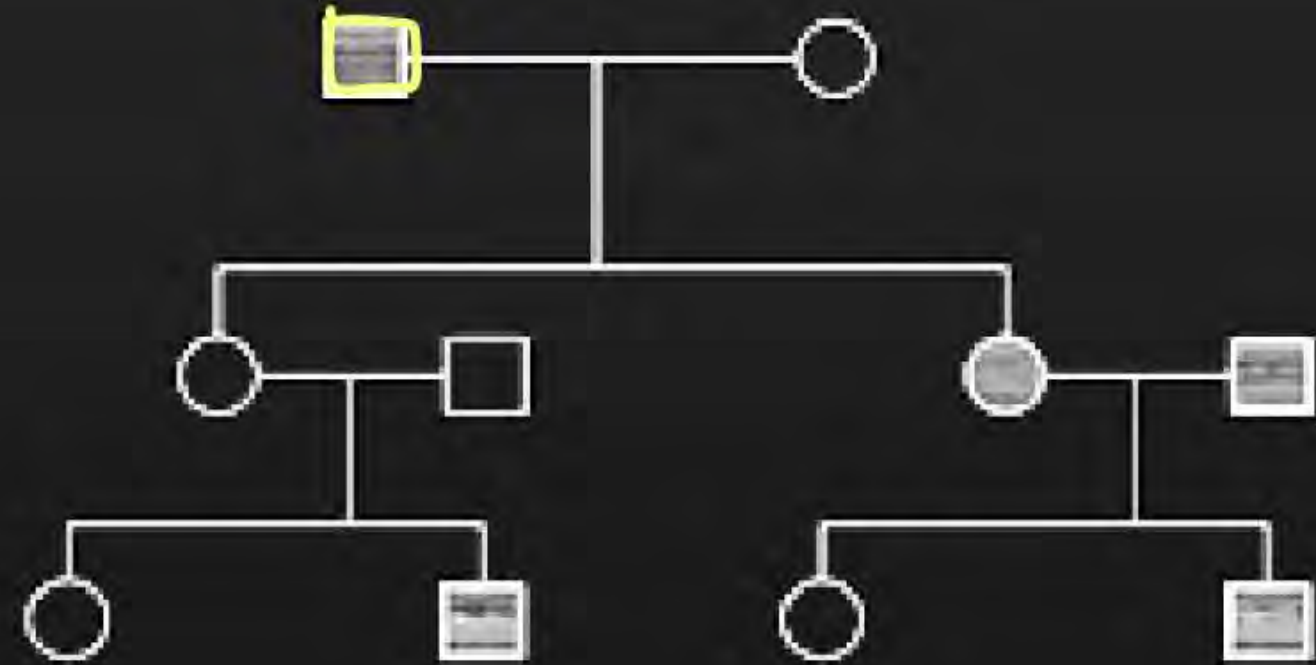
- A** 4 ✓
- B** 8
- C** 12
- D** 16

$$2^n = 4$$

Question

The given pedigree does not represent inheritance of _ disease

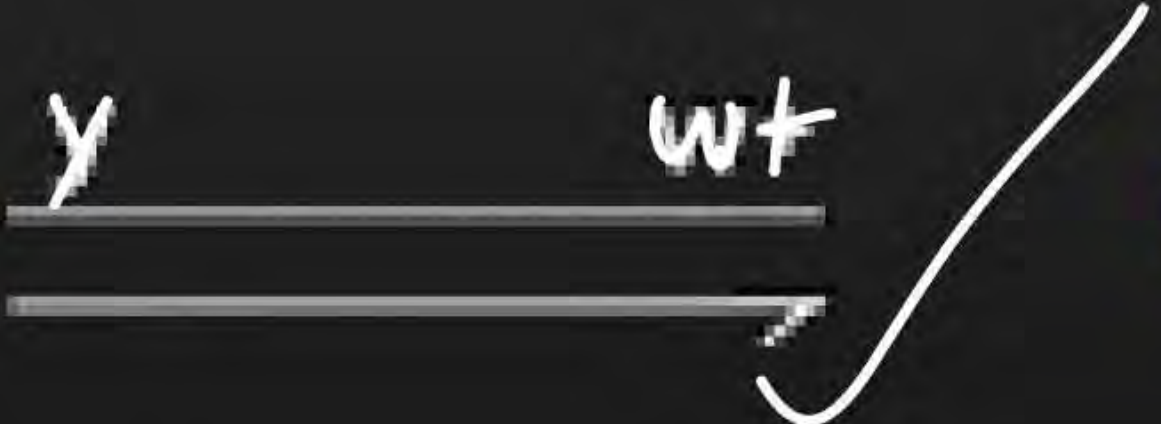

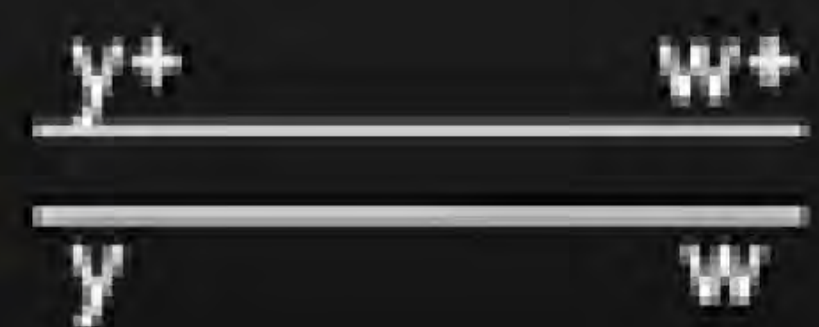

- A** Colour blindness
- B** Thalassemia
- C** Hypertrichosis - *Y linked*
- D** Haemophilia



Question



Which of the following is a recombinant type of progeny obtained by Morgan while carrying out dihybrid cross?

- A** 
- B** 
- C** 
- D** 

Question



Read the given statements and choose correct option

- (A) Turner's syndrome is caused due to the absence of one of the Xchromosome in human female i.e $44+X0$ ✓ = 45
- (B) Down's syndrome is caused due to the trisomic of one of the Xchromosome in human male
- (C) Hypertrichosis is sex-limited trait × *influenced*
- (D) Haemophilia show lethality in human female ✓

A All are correct except (B)

B Only (A) and (D) are correct ✓















C All are correct except (C)

D All are correct



Mendel's law of inheritance

Mendel studied **7 pairs of contrasting characters** in ***Pisum sativum*** - for **7 years 1856 - 1863**. He performed continuous **cross breeding** followed by self pollination and observed the result for several generations. In **total 14 true breeding pea varieties** were selected

Character	Dominant trait	Recessive trait
Seed shape	 Round	 Wrinkled
Seed colour	 Yellow	 Green
Flower colour	 Violet	 White
Pod shape	 Full	 Constricted
Pod colour	 Green	 Yellow
Flower position	 Axial	 Terminal
Stem height	 Tall	 Dwarf



Punnett square -

It was developed by a British geneticist, **Reginald C. Punnett**. It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross.



Inheritance of one gene



pure breeding - homozygous for a character

Same type of gametes

F₁ - phenotype resembles dominant parent

F₁ - heterozygous dominant
Two types of gametes

Only one gene behaviour is studied

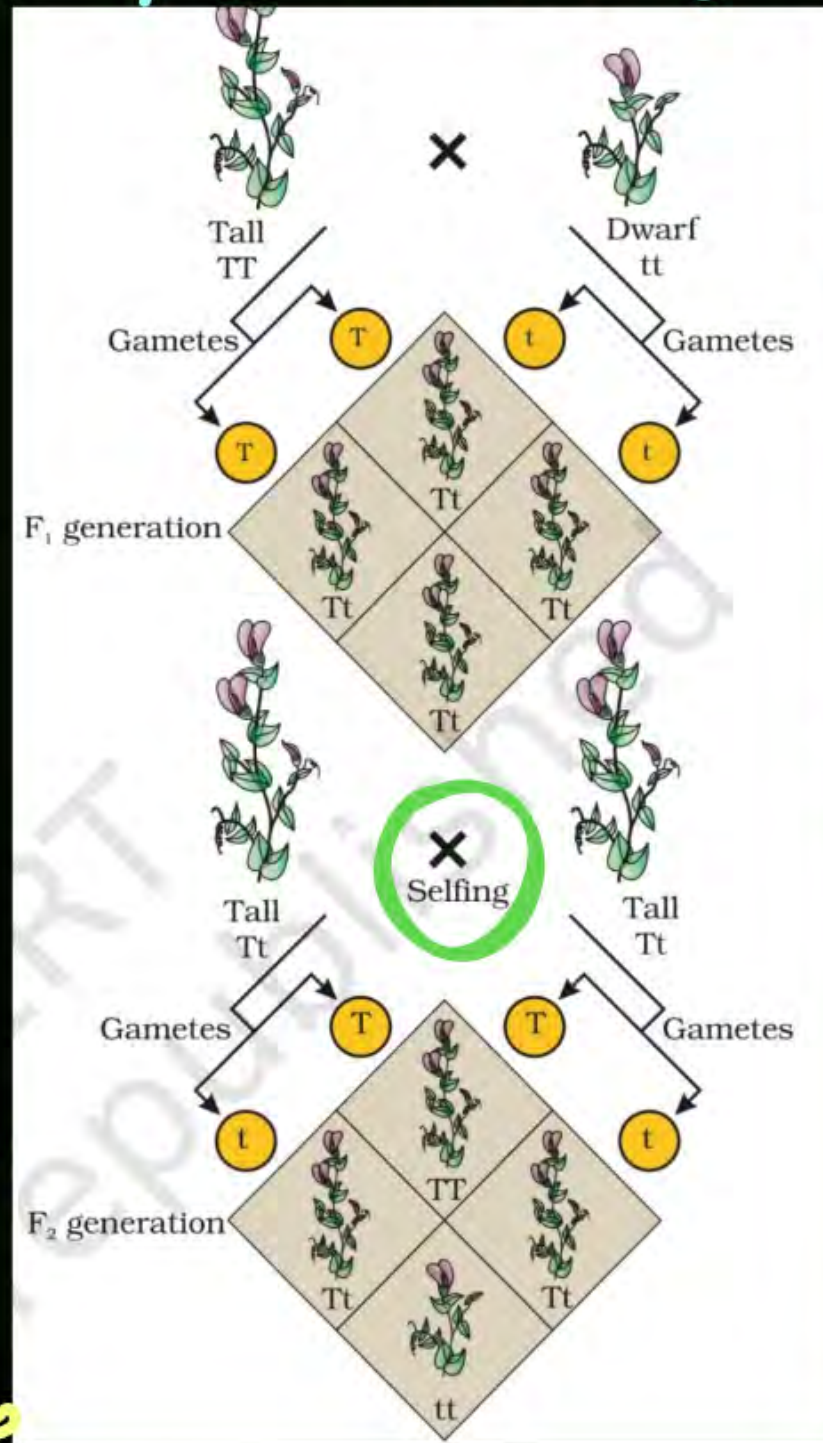
Mendel found that F₁ always resembled dominant parents but in F₂ both traits appeared in 3:1 ratio

There is no intermediate phenotype observed; no blending of character observed

Genotype ratio - 1:2:1 TT:Tt:tt

Phenotype ratio - 3:1 Tall:dwarf

$\frac{1}{4} - TT - \left. \begin{array}{l} \text{Tall} \\ \text{Tall} \end{array} \right\} \frac{3}{4} = 75\%$
 $\frac{1}{2} - Tt - \left. \begin{array}{l} \text{Tall} \\ \text{Tall} \end{array} \right\}$
 $\frac{1}{4} - tt - \text{dwarf} \quad \frac{1}{4} = 25\%$
 homozygous $\frac{1}{2}$
 heterozygous $\frac{1}{2}$





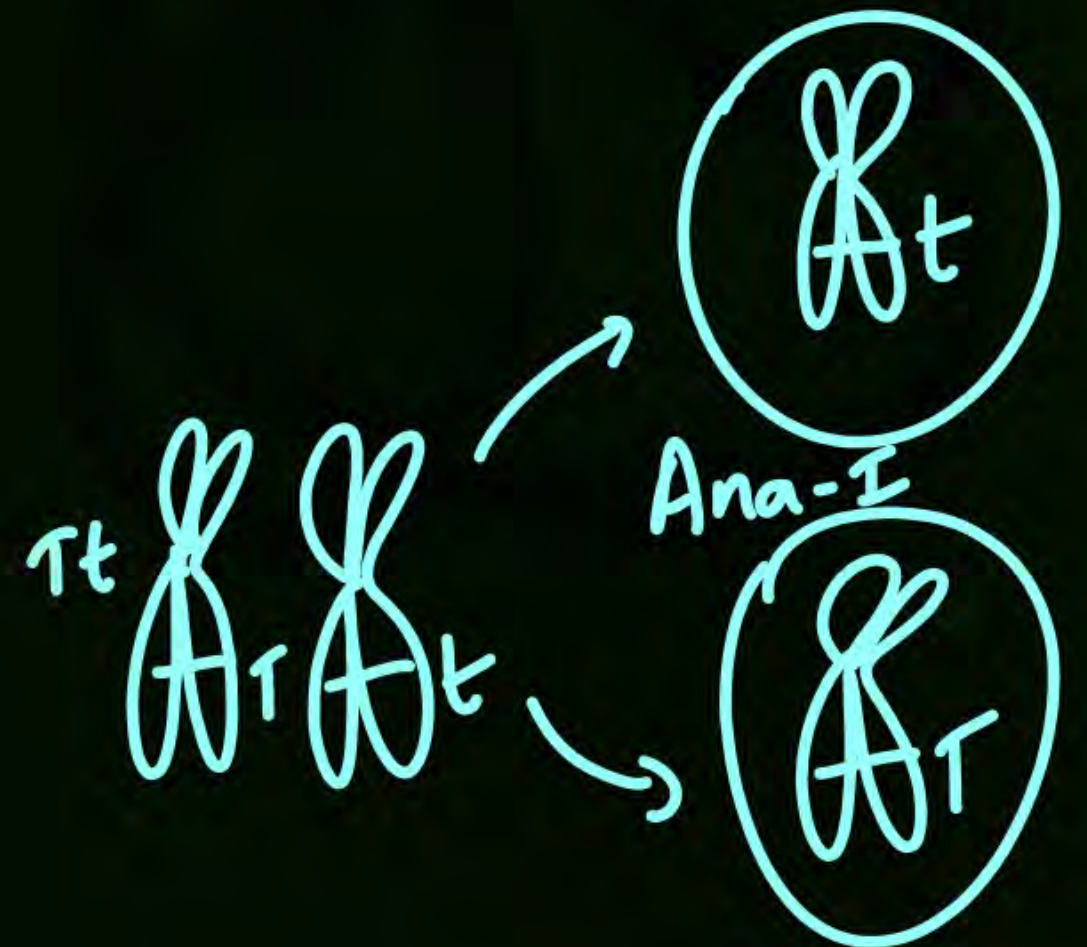
Laws proposed based on monohybrid cross

Law of dominance: not a universal law

- (i) Characters are controlled by discrete units called factors. (genes/alleles)
- (ii) Factors occur in pairs. $\rightarrow 2n$.
- (iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

Law of segregation: universal law

The factors or alleles of a pair segregate from each other such that gametes receive only one of the two factors





TEST CROSS:-

To determine the **genotype of a tall plant at F2**, Mendel crossed the tall plant from F2 with a dwarf plant. This he called a test cross

1:1

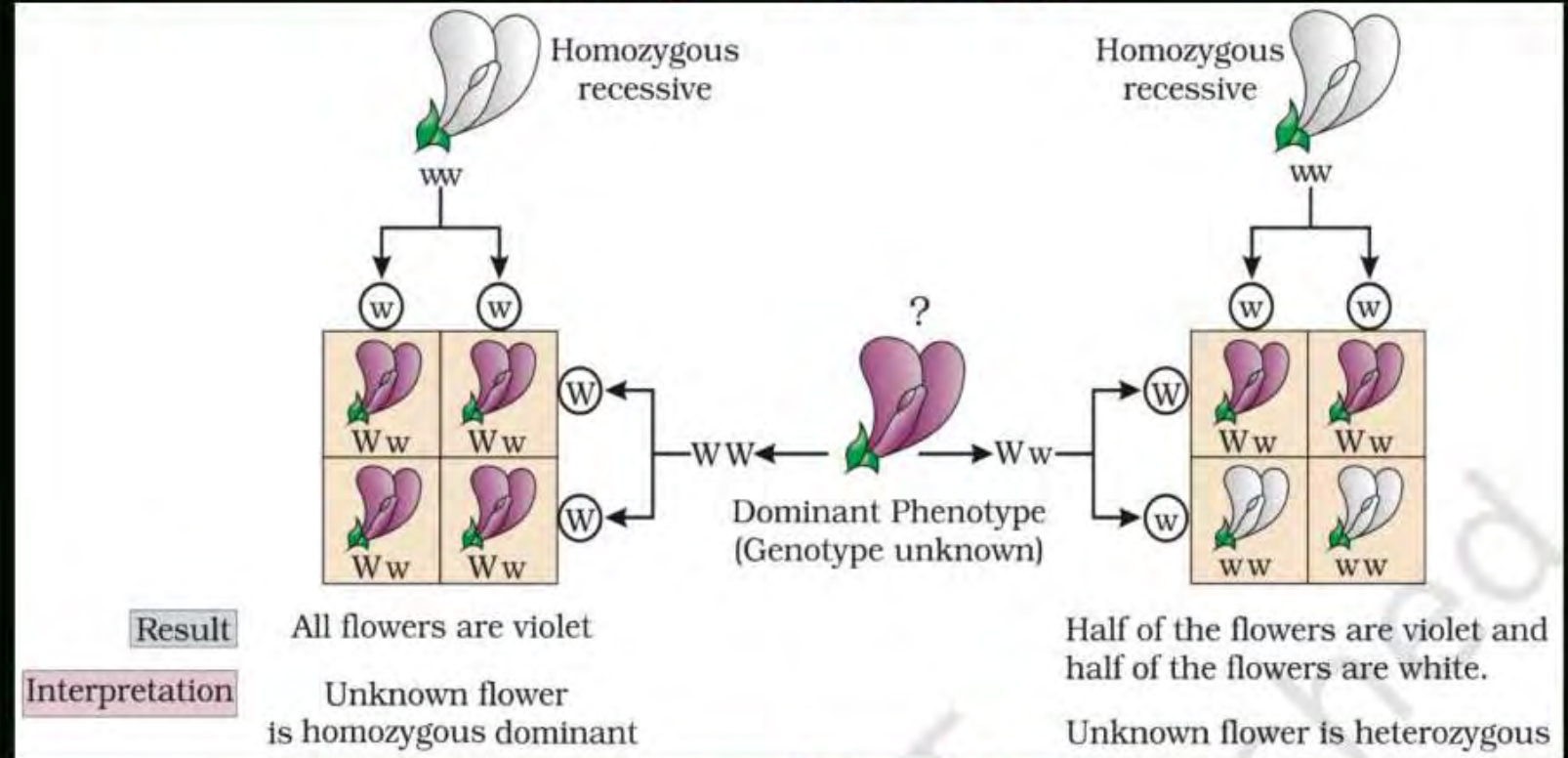
dominant : recessive phenotype

$Ww \times ww$

$(W) (w) (w)$

$Ww \quad ww$

Unknown dominant phenotype
OR genotype $(F_1 \text{ or } F_2)$ \times recessive phenotype
 $F_1 \times$ recessive OR $Tt \times tt$



Unknown? $WW \times ww$

$(w) (w)$

$Ww - 100\% \text{ violet}$



deviation from Mendelism

Incomplete dominance eg Snapdragon, Antirrhinum sp.

Exception to Mendel's observation

In a **heterozygous condition** the **dominant allele** does **not express itself completely** producing an **intermediate phenotype** which does **not resemble** either of the parent in F1 generation.

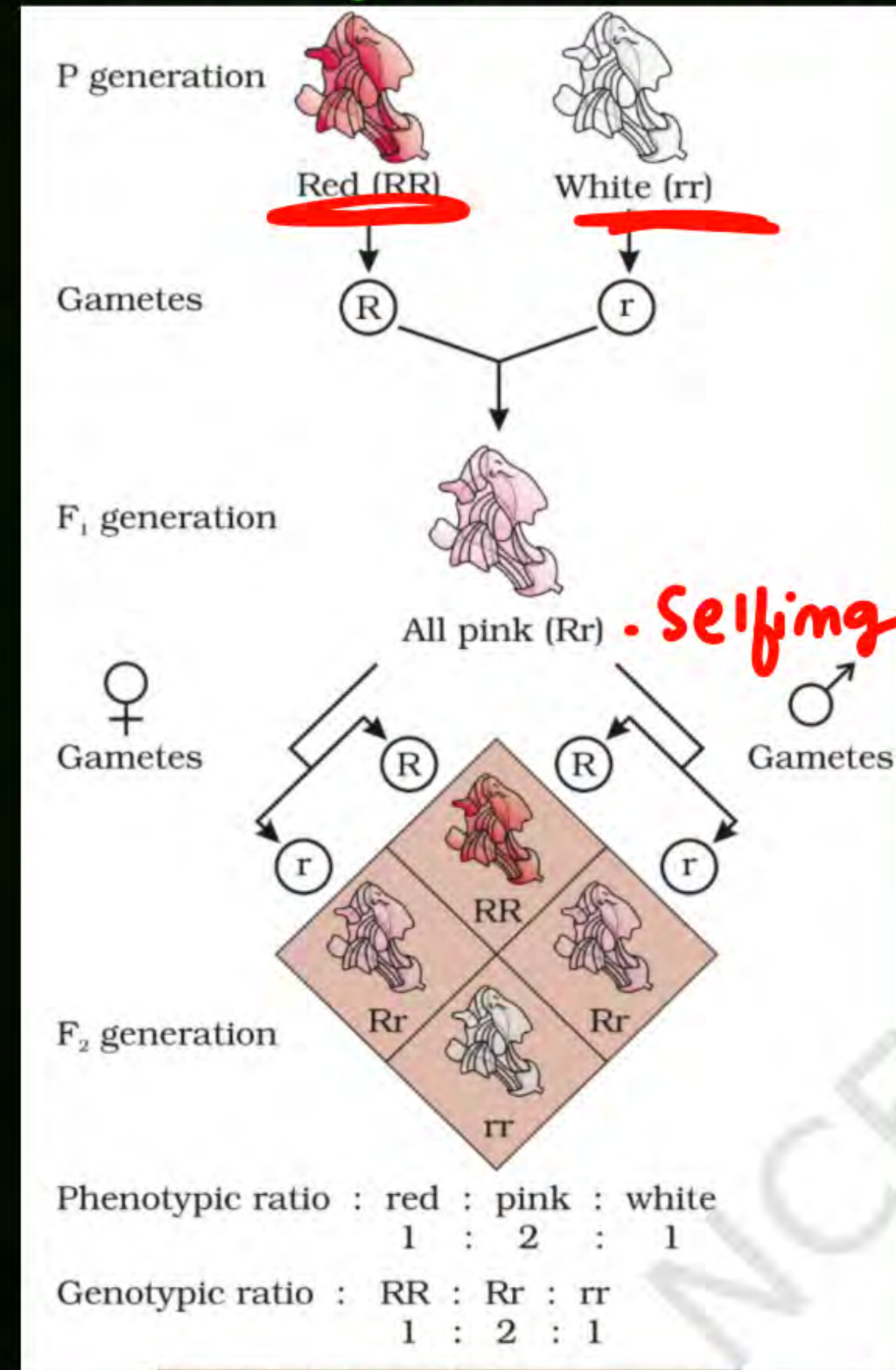
Genotype ratio - 1:2:1

Phenotype ratio - **1:2:1** ; **red: pink: white**

3:1
1:2:1

RR Rr rr

Blending of alleles or characters





Concept of dominance

Rg

In a diploid organism, there are two copies of each gene, i.e., as a pair of alleles.



Let us assume (as is more common) that the normal allele produces the normal enzyme that is needed for the transformation of a substrate S. Theoretically, the modified allele could be responsible for production of –

(i) the normal/less efficient enzyme, the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait → **dominant trait**

(ii) a non-functional enzyme, or
(iii) no enzyme at all

if the allele produces a non-functional enzyme or no enzyme, the phenotype may be effected, this is **recessive trait**.



The **unmodified (functioning)** allele, which represents the original phenotype is the **dominant allele** and the **modified allele** is generally the **recessive allele**

$I^A I^B i \rightarrow$ multiple allelism
 $I^A, I^B \text{ \& } i \rightarrow$ dominant-recessive
 $I^A \text{ \& } I^B \rightarrow$ Co-dominance



Co-dominance :

ABO blood grouping in humans,
 Controlled by I^A, I^B, i gene.

I^A, I^B are co-dominant producing AB blood group.

There are 6 genotypes and 4 phenotypes.

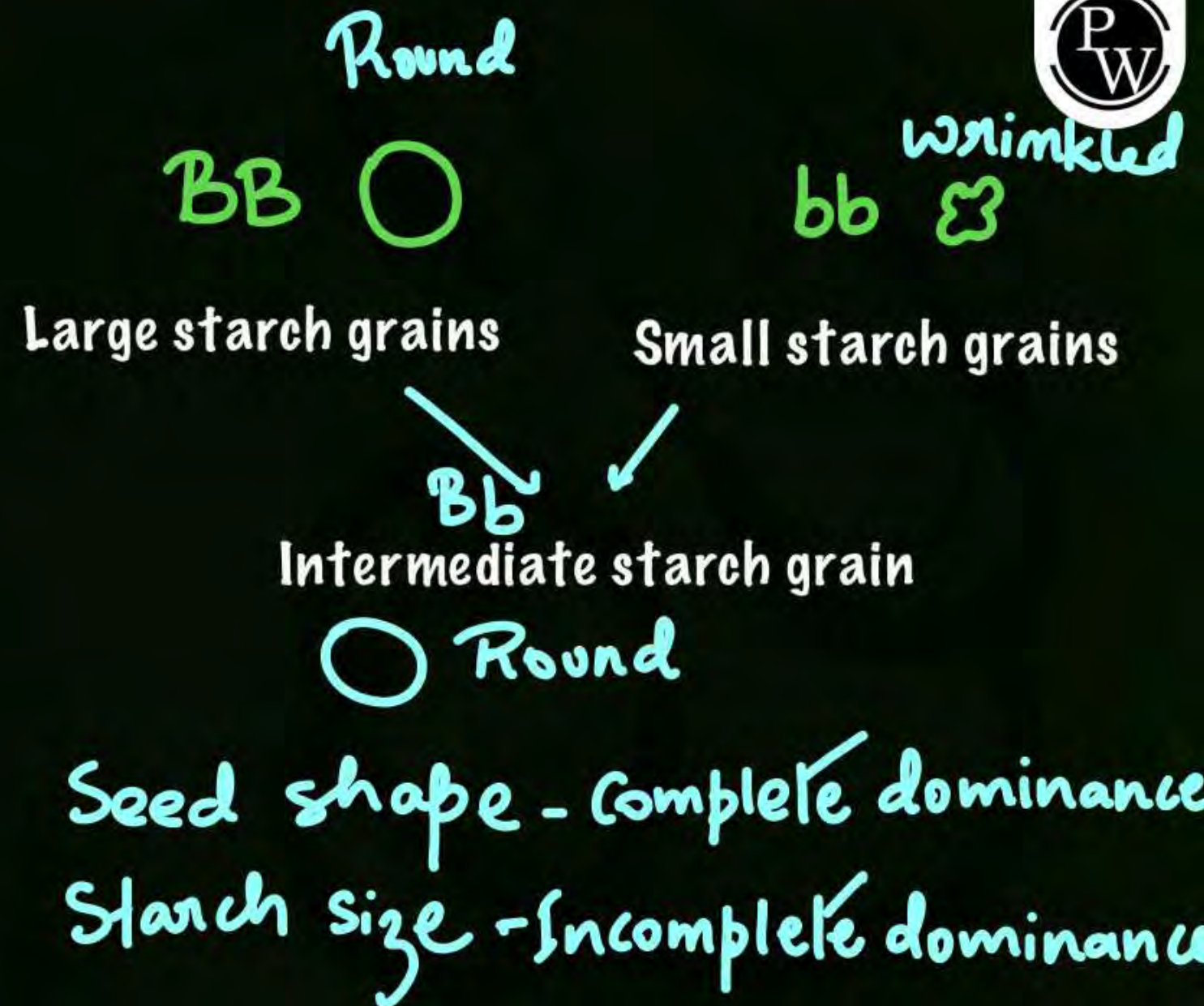
Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	$i i$	O



Pleiotropy - A **single gene** can exhibit **multiple phenotypic expression**.

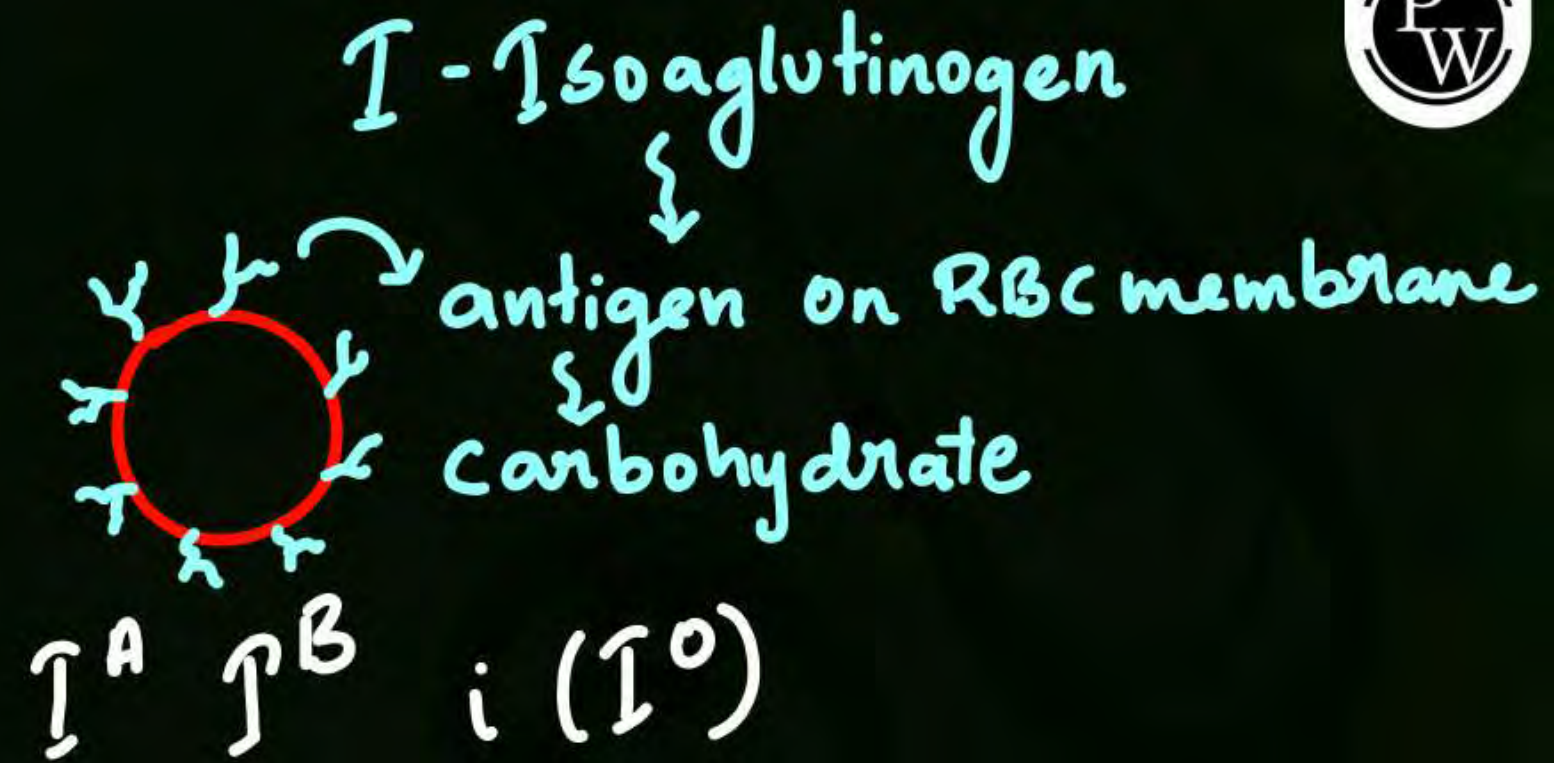
E.g. **Phenylketonuria** caused by gene phenylalanine hydroxylase which causes mental retardation, reduction in hair and skin pigmentation.

Starch synthesis gene in pea - seed shape and size of starch grain get affected
seed shape shows dominance size of starch grain shows incomplete dominance





Multiple alleles -
When **one character is controlled by more than two alleles in a population**
Eq: ABO blood grouping in humans



In an individual only two of the three alleles seen



INHERITANCE OF TWO GENES



- Two genes controlling two different characters are studied

- Phenotypic ratio **9:3:3:1** (Round yellow: round green: wrinkled yellow: wrinkled green) — *Phenotype - 4*

- Genotypic ratio **1:2:2:4:1:2:1:2:1** — *9 genotypes*

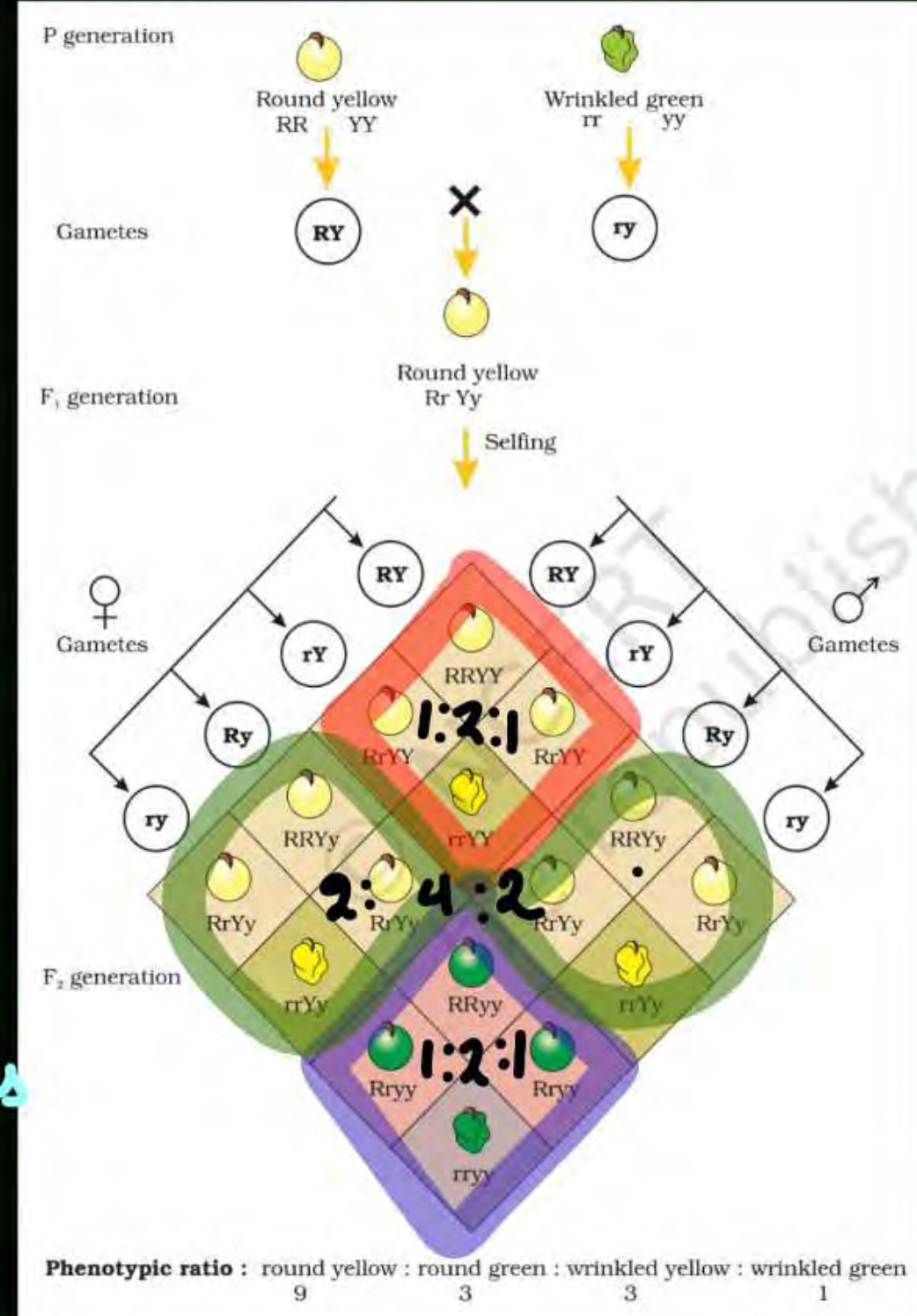
Law based on dihybrid cross

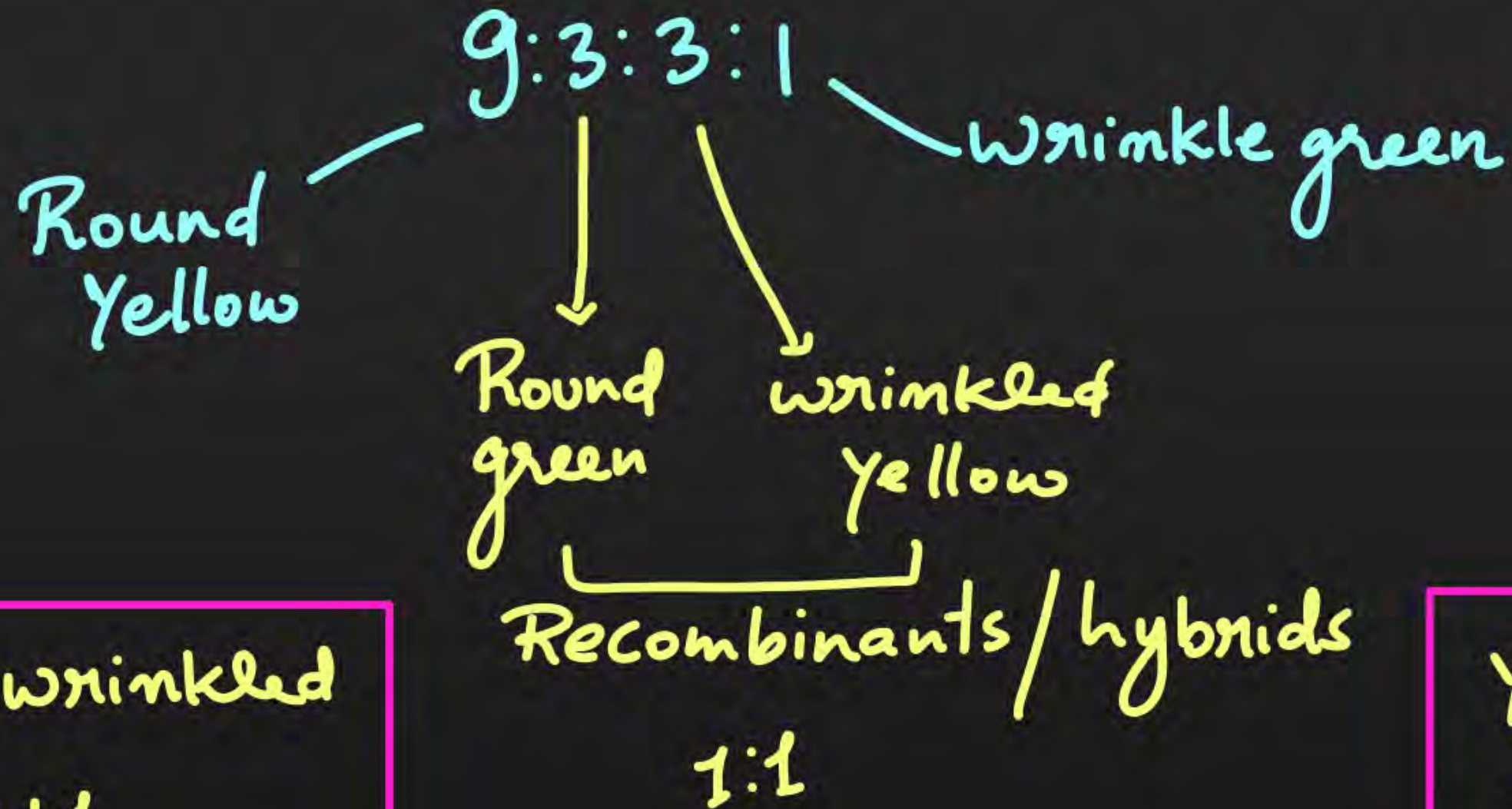
Law of independent assortment

When two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters.

not a universal law

not followed by linked genes





Round : wrinkled
 $9+3 : 3+1$
 $12:4$
 $3:1$

Yellow : green
 $9+3 : 3+1$
 $12:4$
 $3:1$

CHROMOSOMAL THEORY OF INHERITANCE



- Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900. **Correns, De Vries, Tshermack.**

Why Mendel's work went unnoticed??

communication was **not easy** (as it is now) in those days and his work could not be widely publicised.

his concept of **genes** (or factors, in Mendel's words) as **stable and discrete** units that controlled the expression of traits and, of the pair of alleles which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.

Mendel's approach of **using mathematics** to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.

though Mendel's work suggested that **factors (genes)** were discrete units, he could not provide any physical proof for the existence of factors or say what they were made of.

Walter Sutton and Theodore Boveri it's that behaviour of chromosomes was parallel to behaviour of genes and they used chromosome movement to explain Mendel's laws. Experimental verification was done by T.H. Morgan who worked with fruit flies *Drosophila melanogaster*.

Drosophila was suitable for genetic studies

- They could be grown on simple synthetic medium in the laboratory.
- They complete their life cycle in about two weeks, and a single mating could produce a large number of progeny flies.
- Also, there was a clear differentiation of the sexes – the male and female flies are easily distinguishable.
- Also, it has many types of hereditary variations that can be seen with low power microscopes.

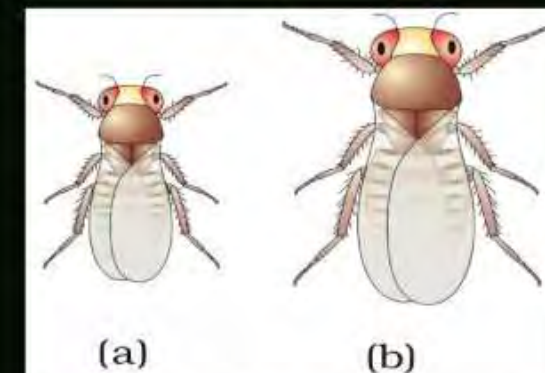


Figure 5.10 *Drosophila melanogaster* (a) Male

Linkage and recombination



- Crosses carried out were **dihybrid crosses**
- Morgan hybridised yellow-bodied, white-eyed females to brown-bodied, red-eyed males and intercrossed their F1 progeny. He observed that the two genes did not segregate independently of each other and the F2 ratio deviated very significantly from the 9:3:3:1 ratio.
- The genes were located on the X chromosome and it was seen that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations were much higher than the non-parental type.
- Linkage:- refers to physical association of two genes located on same chromosome





Cross A

Body colour - Brown, Yellow
eye colour - Red, white

Cross B

eye colour
wing size - normal
miniature

Parental type (98.7%)
↓
Recombinant types (1.3%)

Parental type (62.8%)
↓
Recombinant types (37.2%)

Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript
Note: The strength of linkage between y and w is higher than w and m.

On X¹ Chromosome

Recombinant % low = tightly linked genes

dominant/wild y^+ y = mutant/recessive

Brown body

Yellow body

Genetic mapping - Alfred Sturtevant



Morgan's student Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and 'mapped' their position on the chromosome.

Genetic maps are extensively used as a starting point in the sequencing of whole genomes

1% recombination: 1 map unit = 1 centiMorgan

1.3% = Body colour & eye colour distance = 1.3 mu or 1.3 cM

POLYGENIC INHERITANCE



One phenotype controlled by more than one gene

Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment.

Human skin colour - three genes A, B, C control skin colour in human Height; IQ humans.

The dominant forms A, B and C responsible for dark skin colour

The recessive forms a, b and c for light skin colour.

The genotype with all the dominant alleles (AABBCC) will have the darkest skin colour and that with all the recessive alleles (aabbcc) will have the lightest skin colour.

In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive. more the no. of dominant alleles = darker the skin

SEX DETERMINATION

Henking (1891) could trace a specific nuclear structure all through spermatogenesis in a few insects

Henking gave a name to this structure as the **X body**

Barren body.

Types of sex determination

XX-XY type - Male heterogamety E.g. **humans, drosophila** - no. of chromosomes same for *Female homogamety*

XX-XO type - Male heterogamety E.g. **Grasshopper** - males have *♂ & ♀ one less chromosome than ♀.*

Z-ZW type - Female heterogamety E.g. Birds

male homogamety
haplodiploid

Sex Determination in Honey Bee

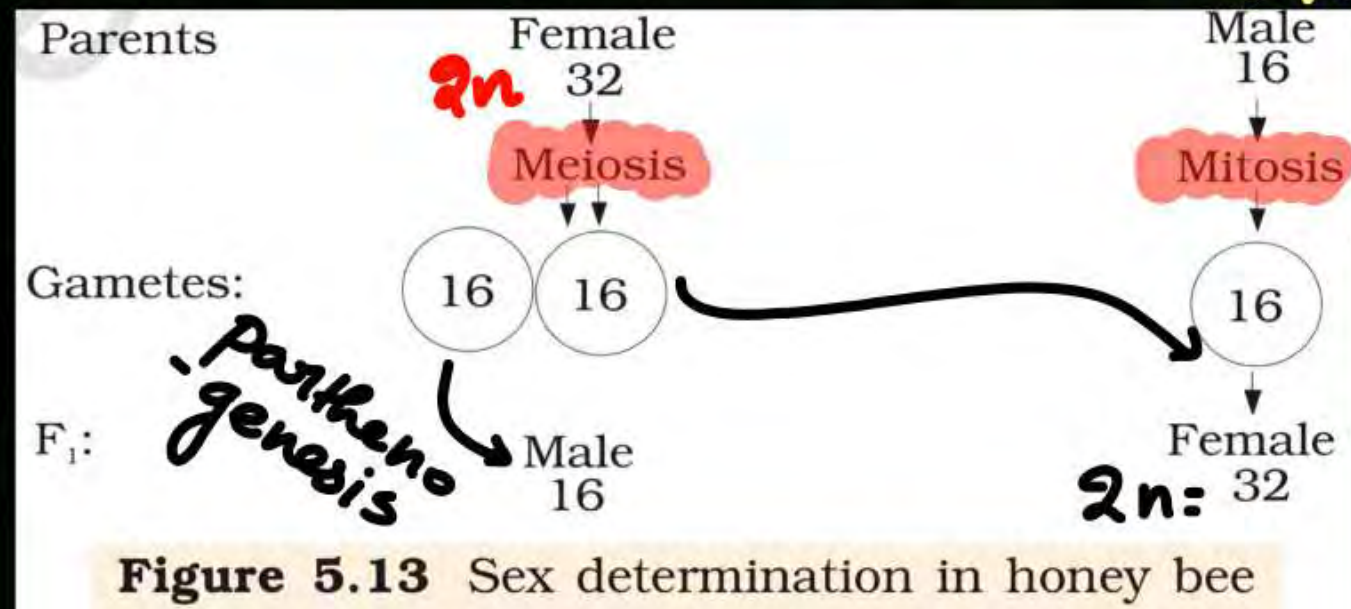


Figure 5.13 Sex determination in honey bee

males do not have father but have grandfather
males → cannot have sons but can have grandsons

MUTATION

Mutation is a phenomenon which results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism

Changes in a single base pair of DNA. This is known as point mutation. A classical example of such a mutation is sickle cell anemia. Deletions and insertions of base pairs of DNA, causes frame-shift mutations

There are many chemical and physical factors that induce mutations. These are referred to as mutagens. UV radiations can cause mutations in organisms – it is a mutagen.

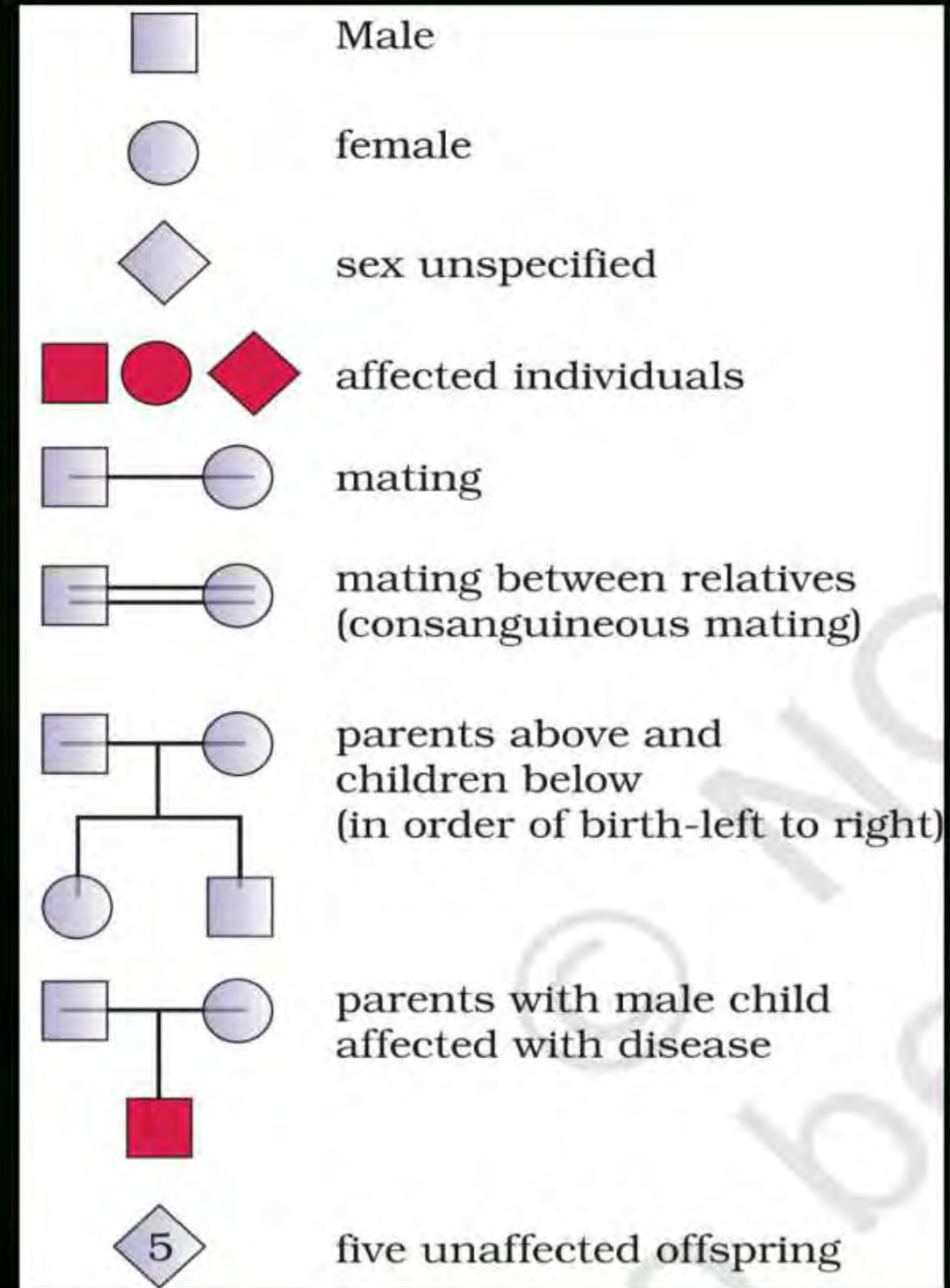
GENETIC DISORDERS

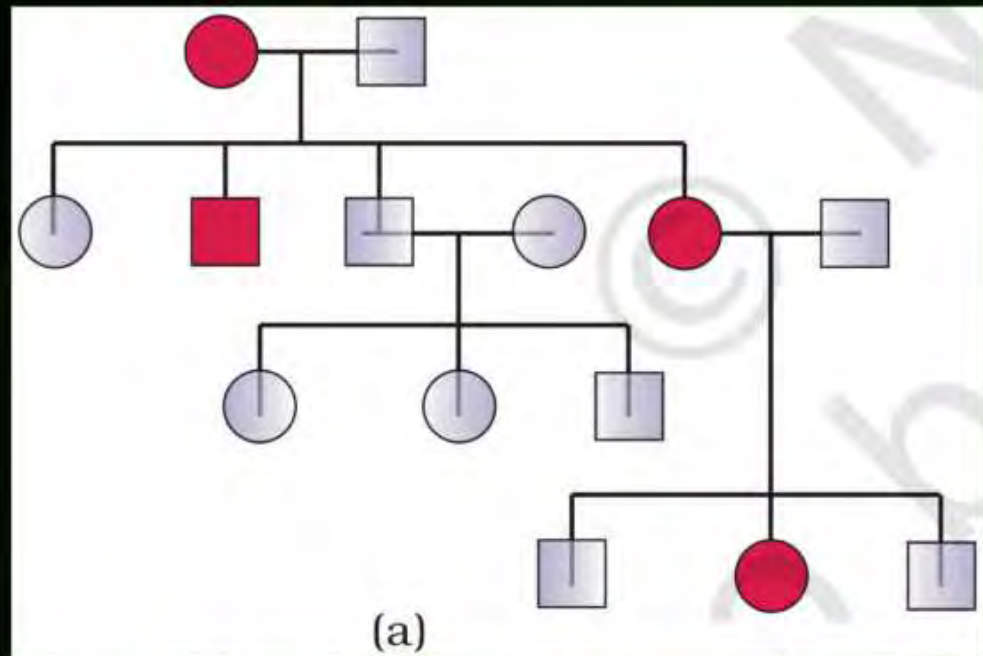
Pedigree Analysis

Analysis of traits in a several of **generations of a family** is called the **pedigree analysis**.

Control crosses are not possible **in case of human beings**, study of the family history about inheritance of a particular trait provides an alternative.

Only for Mendelian disorders

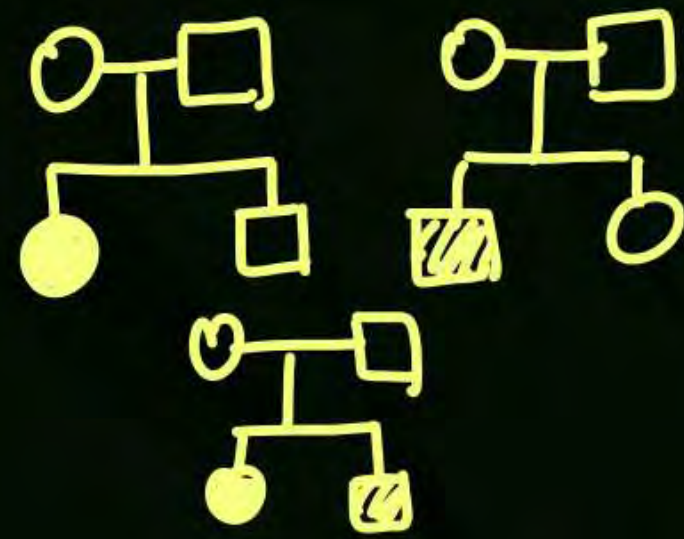




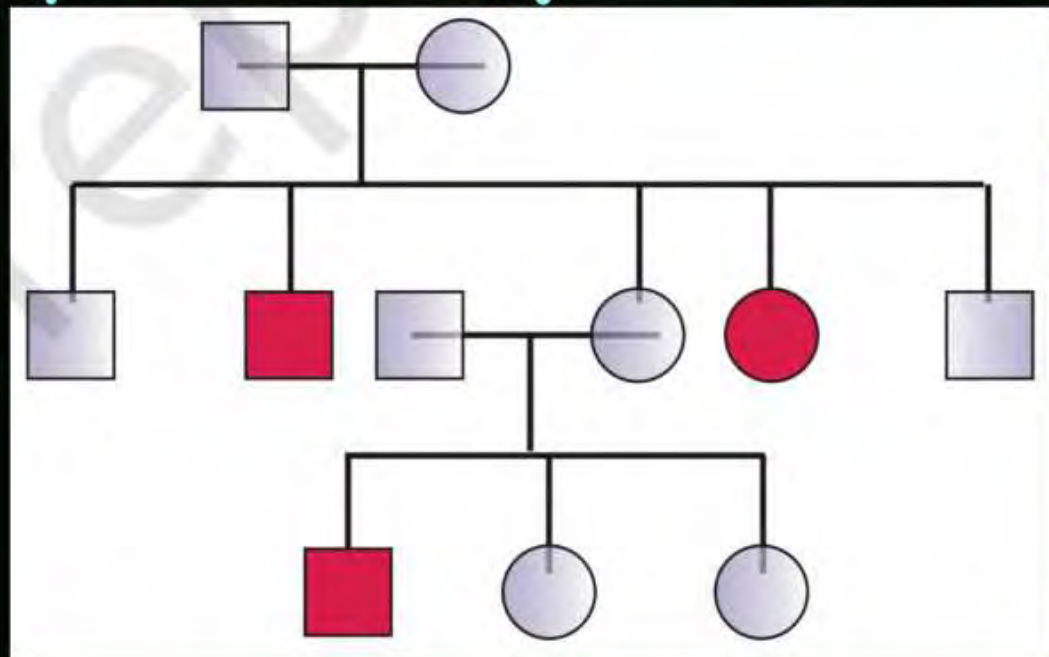
Auto dominant
Myotonic dystrophy

Checklist ✓

a) Domi/Recessive -



b) affected ♀ search
assume it is X-linked
if her father is affected then → X-linked



Auto recessive

if father not affected then auto linked.

Mendelian Disorders



Mendelian disorders are mainly determined by **alteration or mutation in the single gene**. These disorders are transmitted to the offspring on the same lines as we have studied in the principle of inheritance.

The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis.

Most common and prevalent Mendelian disorders are **Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalassaemia, etc.**

Colour Blindness :

It is a **sex-linked recessive disorder** due to defect in either **red or green cone of eye** resulting in failure to discriminate between red and green colour.

Mutation in certain genes present in the **X chromosome.**

→ single 'x'

It occurs in about **8 per cent of males** and only about **0.4 per cent of females**. This is because the genes that lead to red-green colour blindness are on the X chromosome.

Males have only one X chromosome and females have two. The son of a woman who carries the gene has a **50 per cent chance of being colour blind**

Haemophilia

This **sex linked recessive disease**, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied.

In this disease, a **single protein** that is a part of the **cascade of proteins involved in the clotting of blood** is affected.

Due to this, in an affected individual a simple cut will result in non-stop bleeding. The heterozygous female (carrier) for haemophilia may transmit the disease to sons

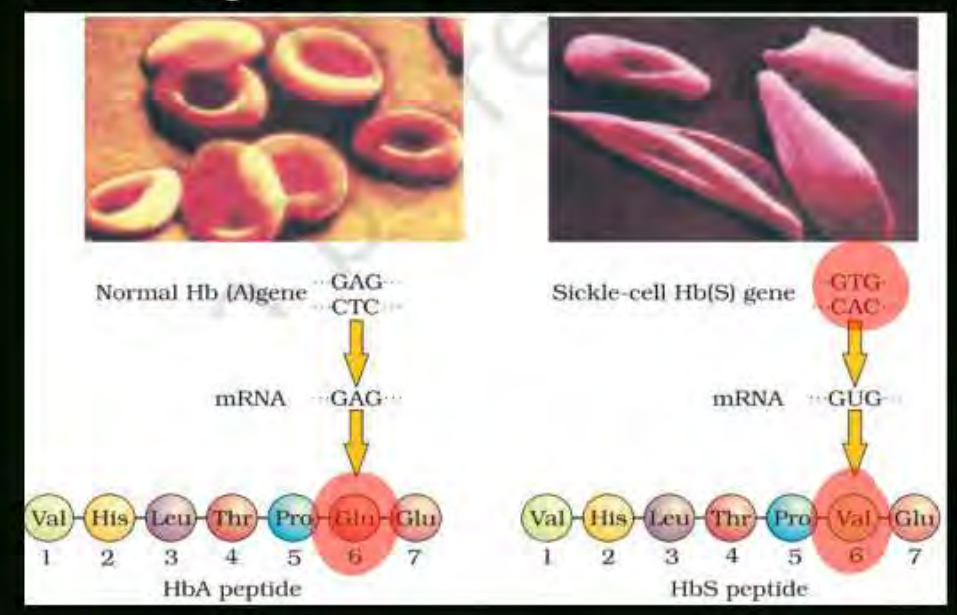
The family pedigree of **Queen Victoria shows a** number of haemophilic descendants as she was a carrier of the disease.

Sickle-cell anaemia: **Gene on chromosome-11**
 $Hb^A Hb^A$; $Hb^A Hb^S$; $Hb^S Hb^S$ - affected

This is an autosome linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous).

The disease is controlled by a single pair of allele, Hb^A and Hb^S . Out of the three possible genotypes only homozygous individuals for Hb^S ($Hb^S Hb^S$) show the diseased phenotype.

Heterozygous ($Hb^A Hb^S$) individuals appear apparently unaffected but they are carrier of the disease as there is 50 per cent probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait



The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure



Thank

You