

Genetics → Branch of Science



↓ Inheritance

Process of transmission
of characters from one
generation to another.

* Heredity

Phenomenon of inheritance of
genes

Degree of difference between
parents & progeny / offsprings.

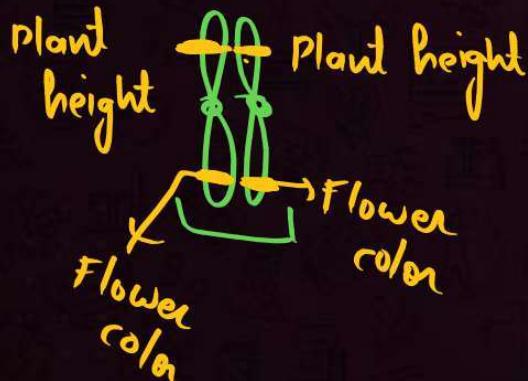
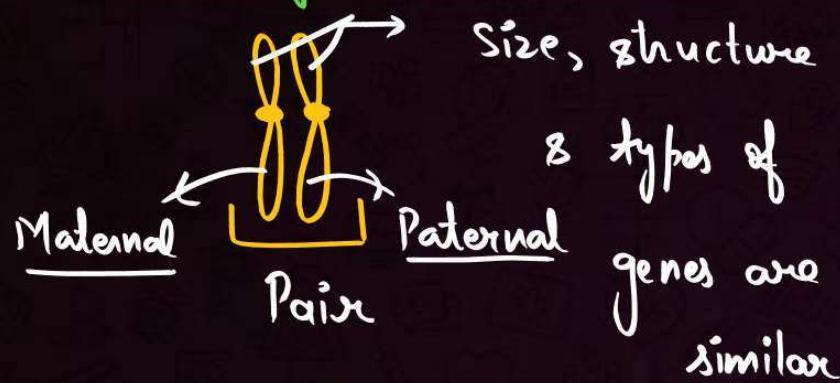
Cause

+ Recombination

↓ Mutation
(Major)

Terminology

Homologous chromosomes



In diploid organism (In diploid cell)

(Chromosomes are in pair (Each chromosome has 2 copies))

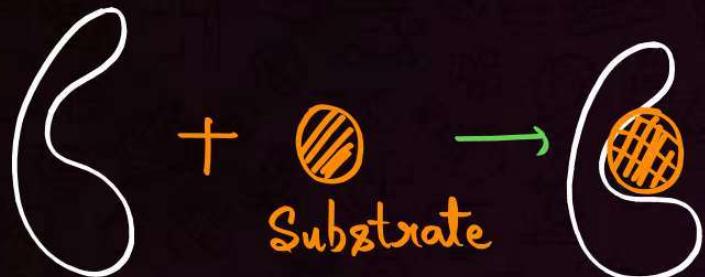
Genes are in pairs (Each gene has 2 copies)

NOTE: In haploid organism | In

Each chromosome has single copy cell

— Genes have single copy

Concept of Dominance



Enzyme
↓

Responsible for
Transformation of
Substrate

==== DNA

"Gene" → codes for information

↓ Transcription

m-RNA

↓ Translation

→ Protein (Act as Enzyme)

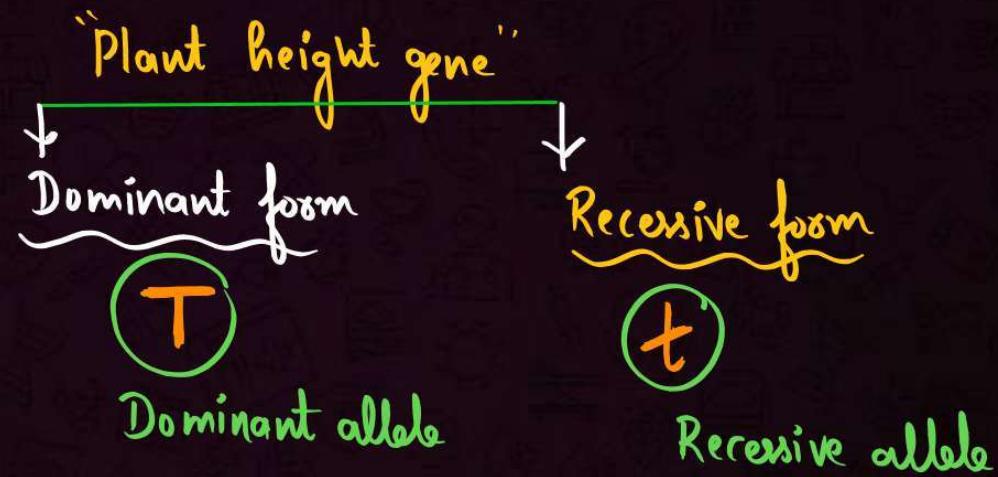
Product
(Responsible
for phenotype
expression).

P
W



Alleles / Allelomorphic pair

Different forms of a same gene.

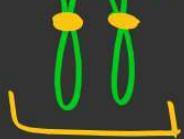


Heterozygous

P·H P·H



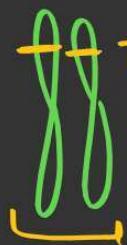
T t



(Tt)

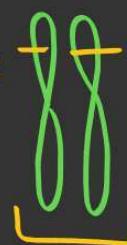
Homozygous

T T



(TT)

t t



(tt)

Phenotype

Morphological appearance
(Feature of an organism)

- * Plant height
- * Flower color

Genotype

Genetic make - up of an organisms.

TT → Genotype

Tt → Genotype

tt → "

*

Character

Feature of an organism

* Flower color

* Seed shape

Trait

Distinguishable form of character

Plant height
character

Tall → Trait

Dwarf → Trait

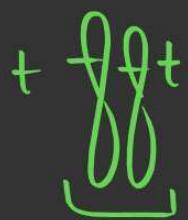
Seed shape → Trait
→ Round → Trait
→ wrinkled → Trait

Pure line / True-breeding Variety

Homozygous for a character / Trait



$\textcircled{\text{I}}\textcircled{\text{T}}$ Pure line



$\textcircled{\text{t}}\textcircled{\text{t}}$ Pure line

Parent lines:

a) Formed by continued Self-Pollination.

b) Shows stable-trait inheritance

c) One only one type of gametes.

Gregor Johann Mendel → Father of Genetics

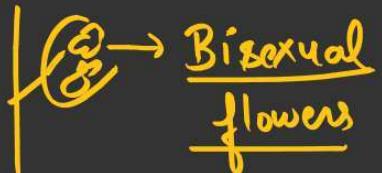
* Born in 1822 Died in → 1884

* Worked on Pisum sativum (Pea plant)

for 7 years (1856-1863) PB

* Work was published in 1865.

Pisum sativum (why)



- a) Short life span
- b) Produce large no. of offsprings / (Seeds)
progeny
- c) Easy to grow in lab.
- d) Has seven pair of contrasting traits
- e) Naturally self-Pollination.
- f) Cross-pollination by Artificial hybridization.

Pisum sativum

Diploid organism

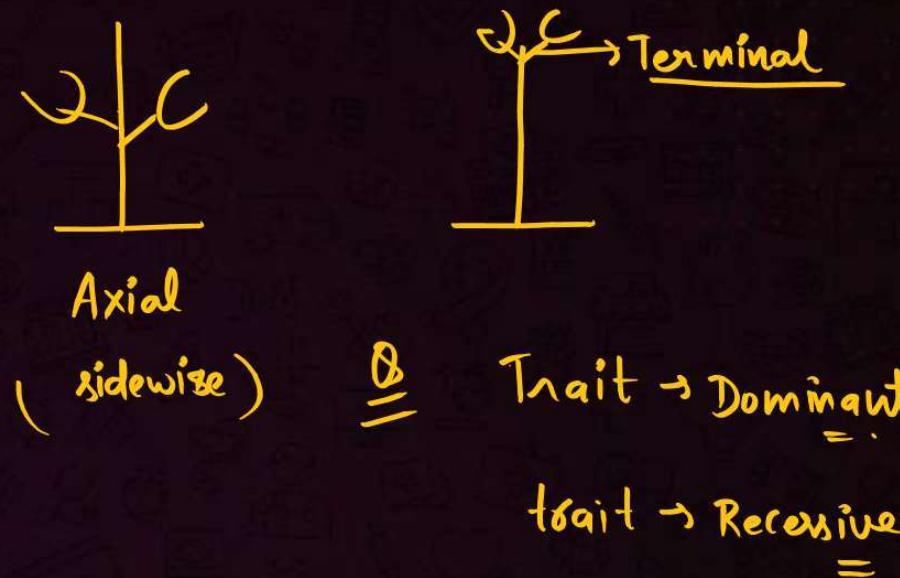
$$2n = 14$$

7 pairs of chromosomes.



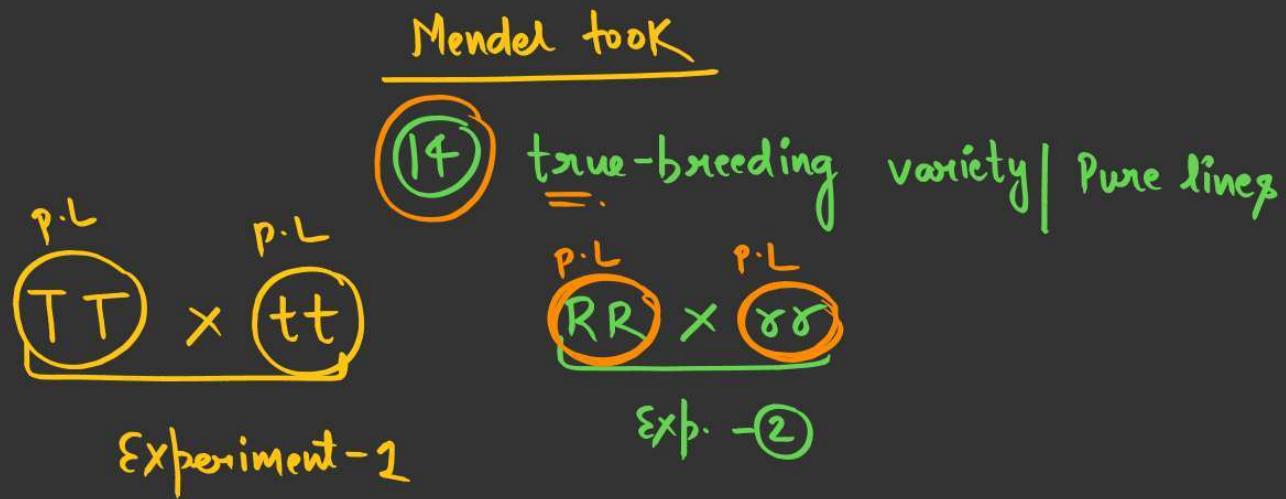
Character	Dominant trait	Recessive trait
Seed shape	Round ✓	Wrinkled ✓
Seed colour	Yellow ✓	Green ✓
Flower colour	Violet Purple	White
Pod shape	Full Inflated	Constricted ✓
Pod colour	Green ✓	Yellow
Flower position	Axial ✓	Terminal ✓
Stem height	Tall	Dwarf ✓

Figure 4.1 Seven pairs of contrasting traits in pea plant studied by Mendel



Mendel was Successful

- ① Mathematical & statistics tools in Biology
(was the first)
- ② He took large sample size



Mendel's work

One-gene inheritance

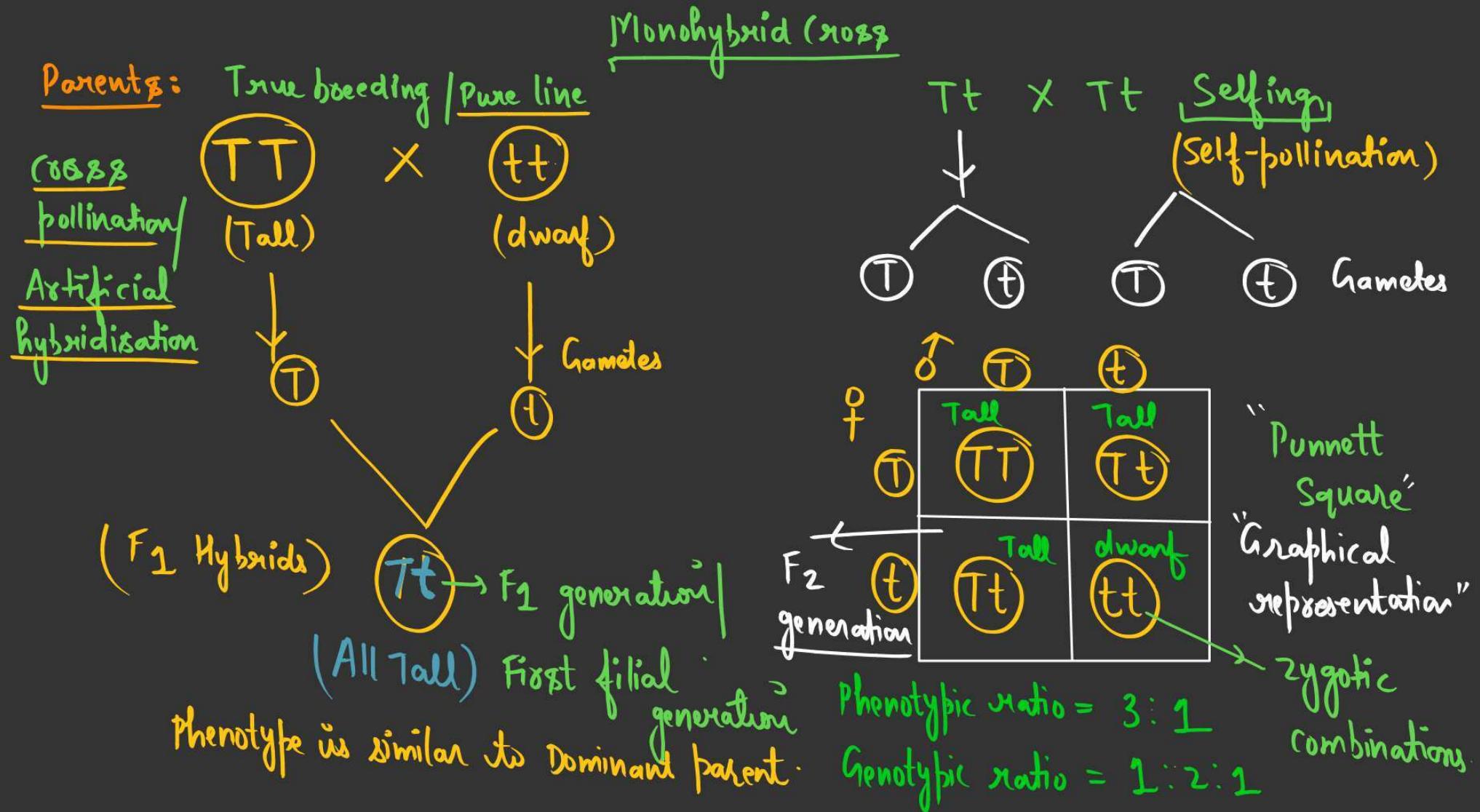
Monohybrid cross

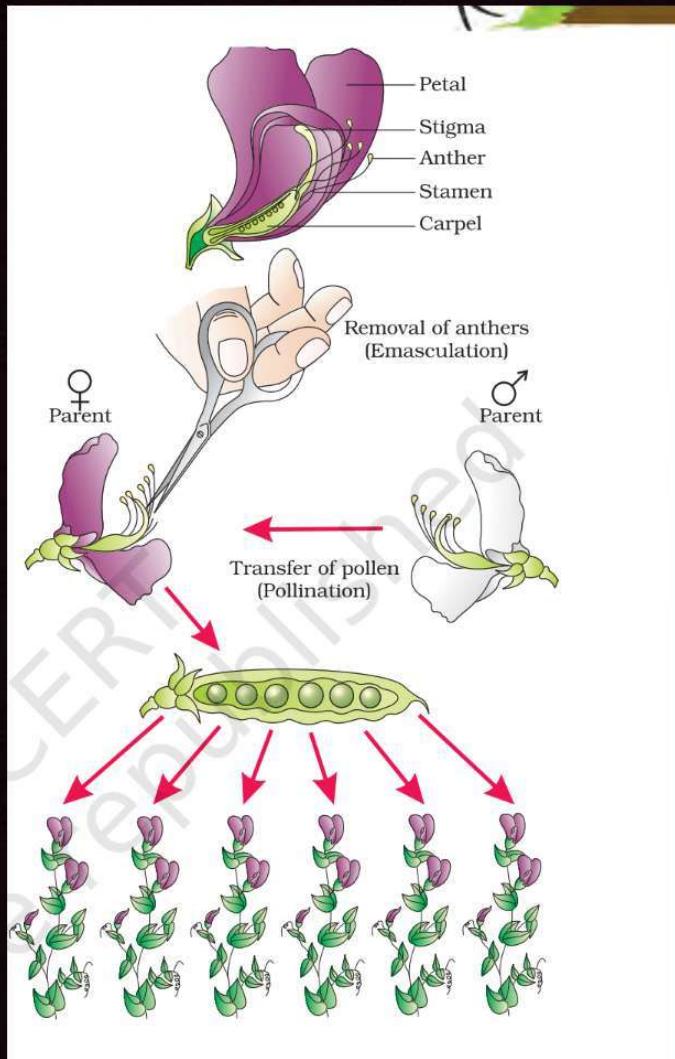
when only inheritance
of only gene is studied

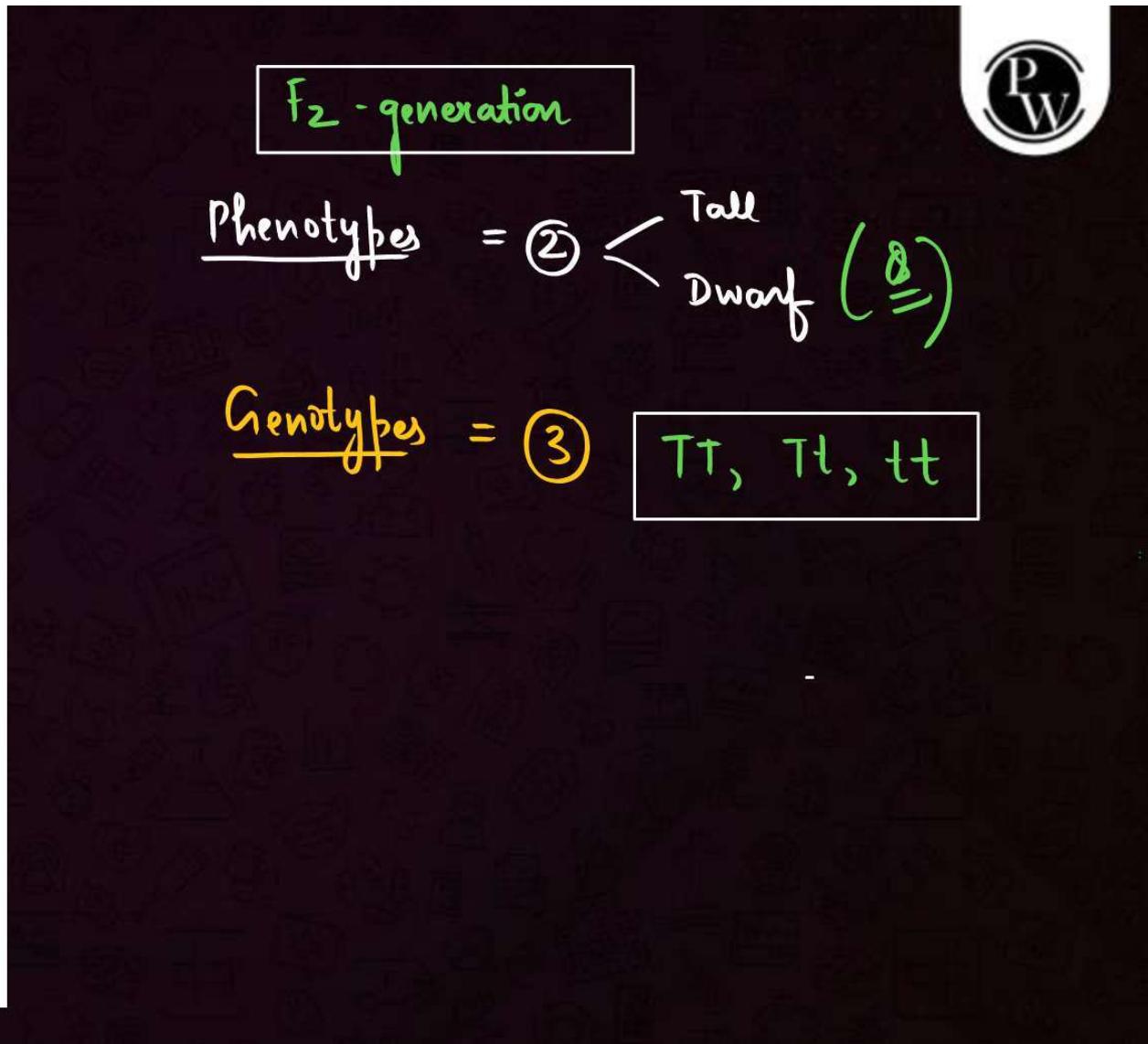
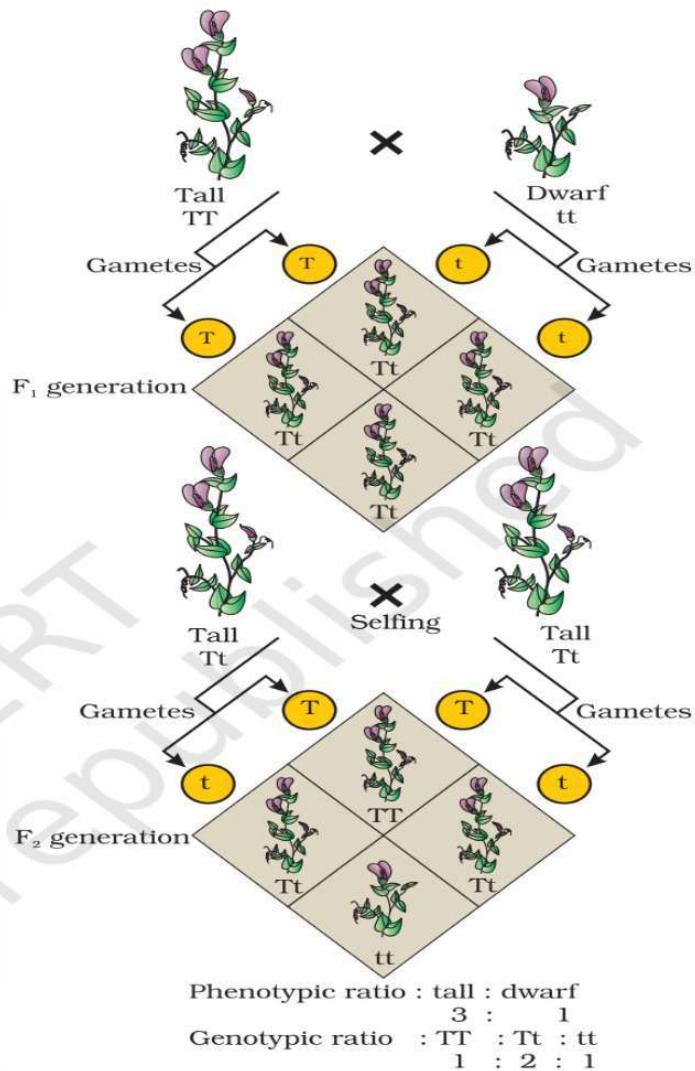
Two-gene inheritance

Dihybrid cross

inheritance of 2 genes
studied together

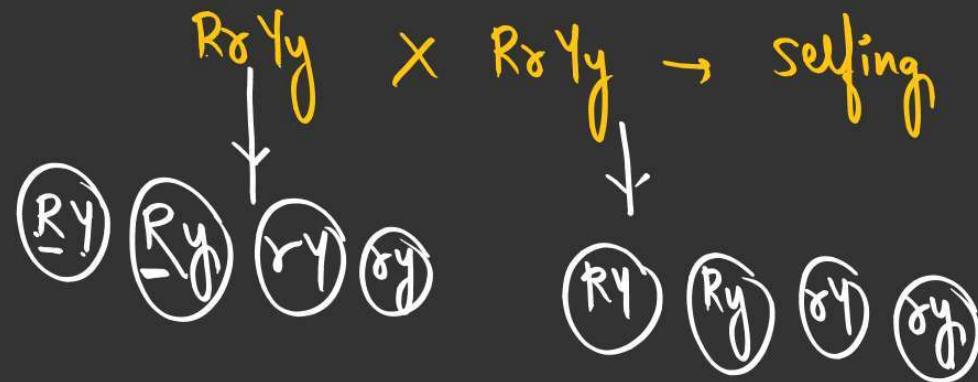
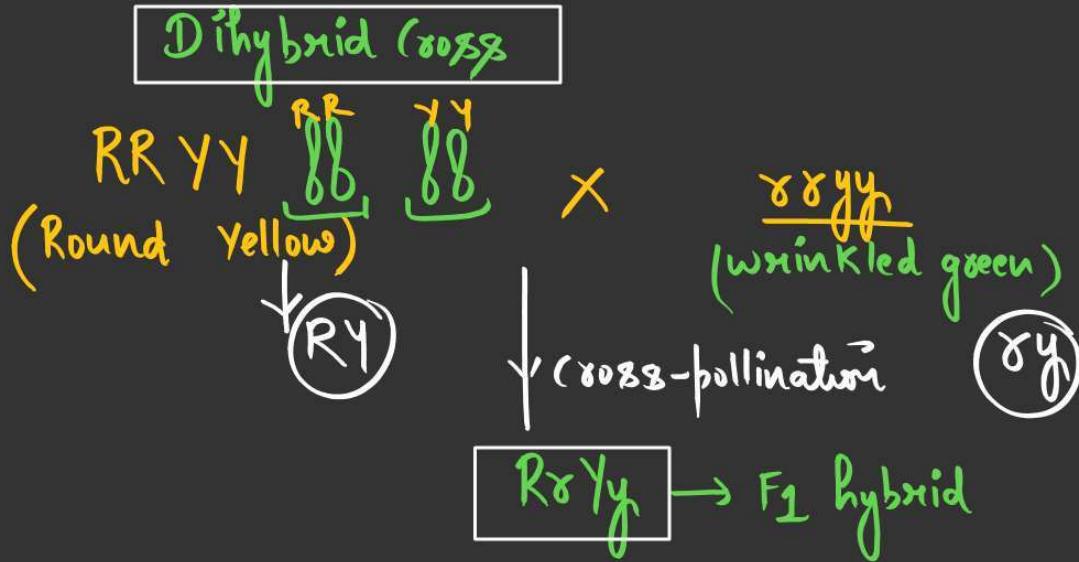


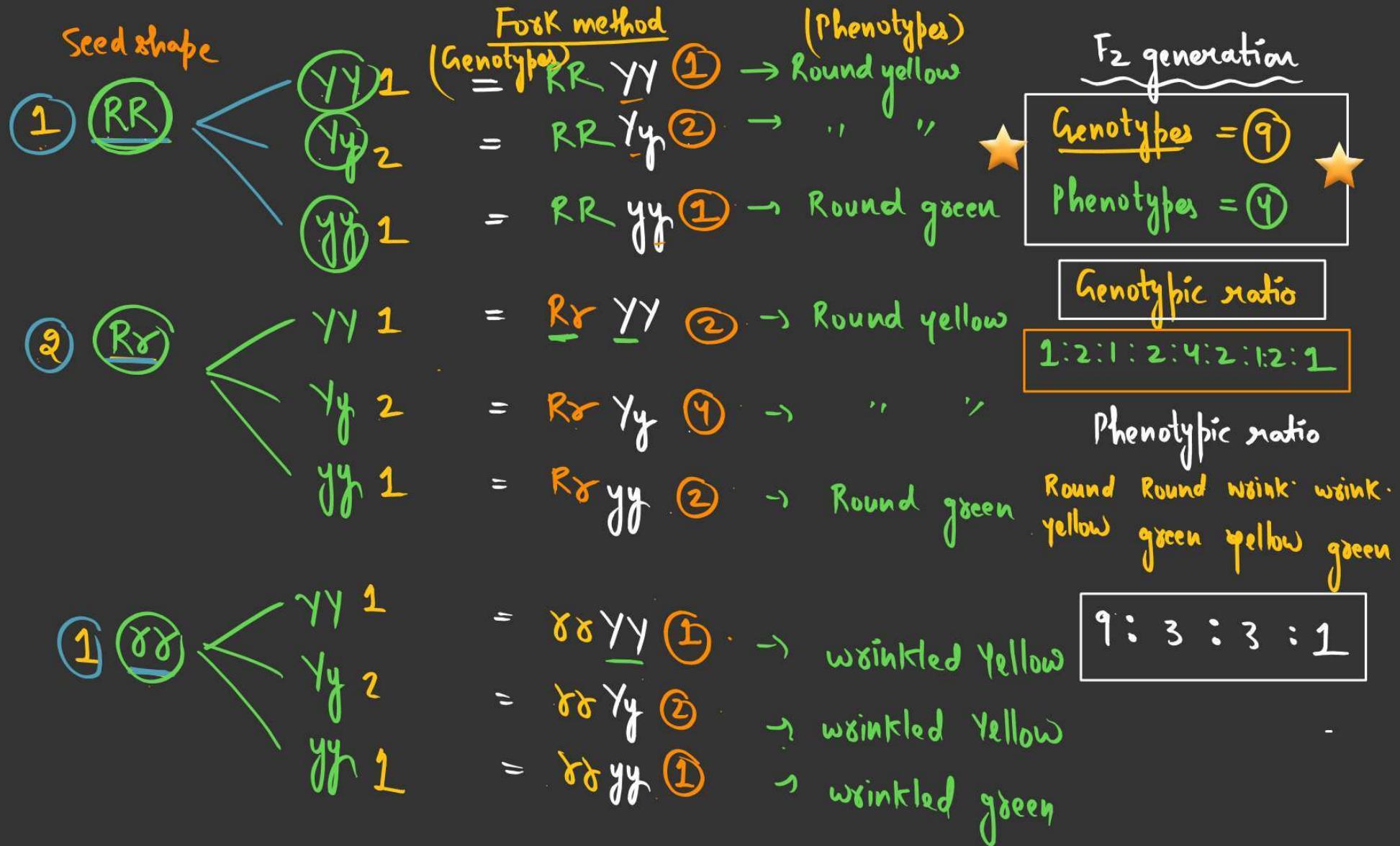





Seed shape
 7th chs.

Seed color
 1st chs.





Laws of inheritance

①

law of DominanceNot Universal

fails at

→ Based on Monohybrid cross.

- * Codominance
- * Incomplete dominance.

4.2.1 Law of Dominance

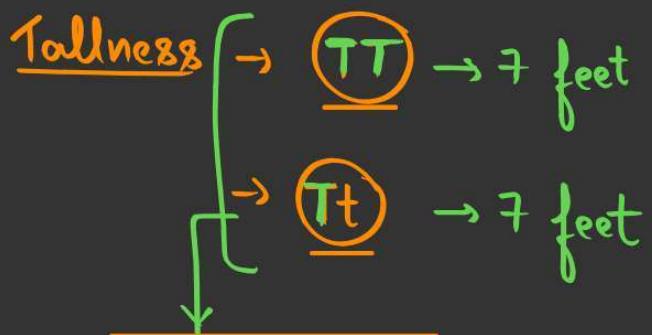
- (i) Characters are controlled by discrete units called factors. ↗ f�+
- (ii) Factors occur in pairs.
- (iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F_1 and the expression of both in the F_2 . It also explains the proportion of 3:1 obtained at the F_2 . 9//

Imp

Dominant trait can express
itself both in heterozygous
& homozygous condition

Recessive trait can
express itself only in
homozygous condition



Dwarfness \Rightarrow tt

Qualitative
inheritance

Q which of the following trait
can express itself only in

Homozygous condition?

Recessive
Trait

- a) Yellow seed $\rightarrow \textcircled{D}$
- b) Axial flower $\rightarrow \textcircled{D}$
- c) ~~Yellow pod~~ $\rightarrow \textcircled{R}$
- d) Violet flower $\rightarrow \textcircled{D}$

Diploid individual

Law of Segregation



Based on : Monohybrid Crosses



Segregation of traits



(Segregation of chromosomes &
genes(factors))

4.2.2 Law of Segregation

8

This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage. Though the parents contain two alleles during gamete formation, the factors or alleles of a pair segregate from each other such that a gamete receives only one of the two factors. Of course, a homozygous parent produces all gametes that are similar while a heterozygous one produces two kinds of gametes each having one allele with equal proportion.





Law of independent Assortment

Based on Dihybrid Cross

Not Universal

fails at "Linkage"

In a dihybrid cross

F₁ hybrid

RyYy

→
Gamete formation

Y
 R
 (1st pair)
 (Seed color)

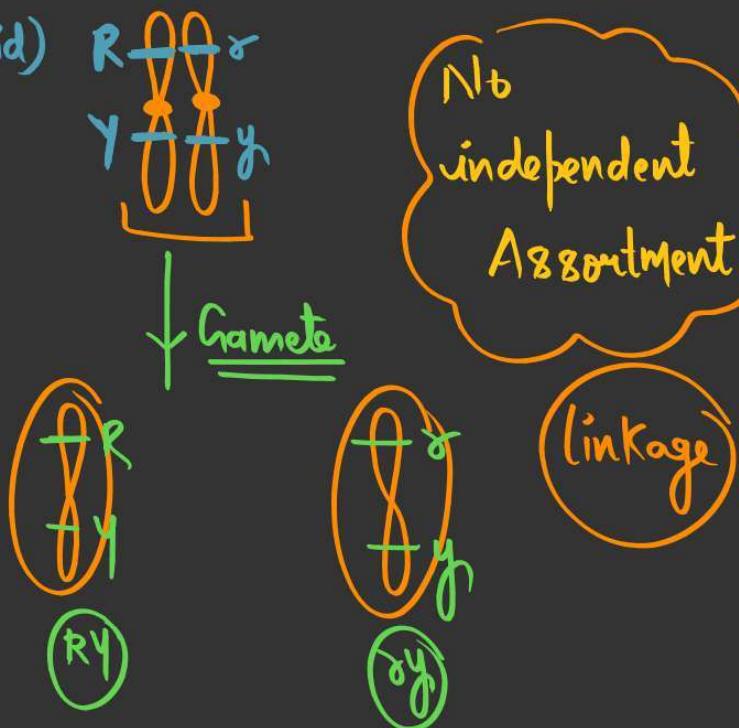
(7th pair)
 (Seed shape)

R
 Y
 RY
 Ry
 25% $\frac{1}{4}$
 25% $\frac{1}{4}$
 25% $\frac{1}{4}$
 25% $\frac{1}{4}$

—

Suppose: These 2 genes were present on same chromosomes.

(F₁ hybrid) R⁺y⁻
(RyYy)



⇒ * NOTE: Independent Assortment can be seen only between genes present on different chromosomes
(Non-homologous chromosomes)

Based upon such observations on **dihybrid crosses** (crosses between plants differing in two traits) Mendel proposed a second set of generalisations that we call Mendel's Law of Independent Assortment. The law states that 'when two pairs of traits are combined in a ~~hybrid~~, segregation of one pair of characters is independent of the other pair of characters'. *



Back-Cross

F₁ hybrid \times Either of the Parent

Dulcossz

F₁ \times Dominant
parent

F₀
ex \rightarrow $(Tt) \times TT$
F₁

"Definition"

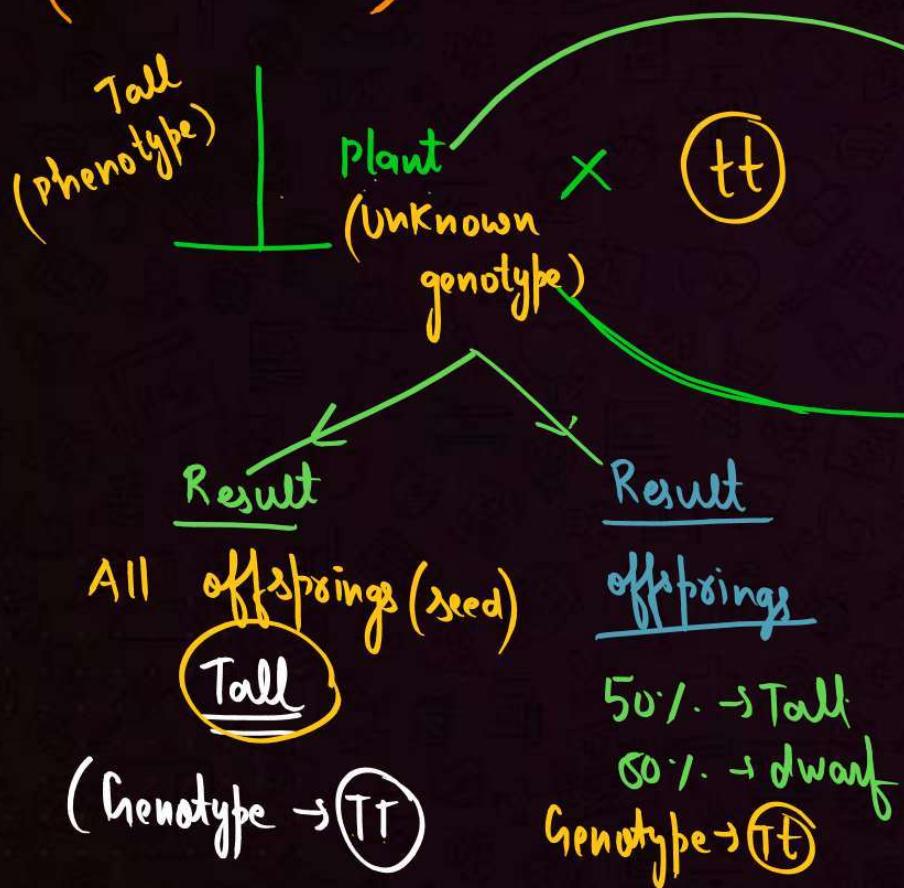
Test Cross

F₁ \times Recessive
parent

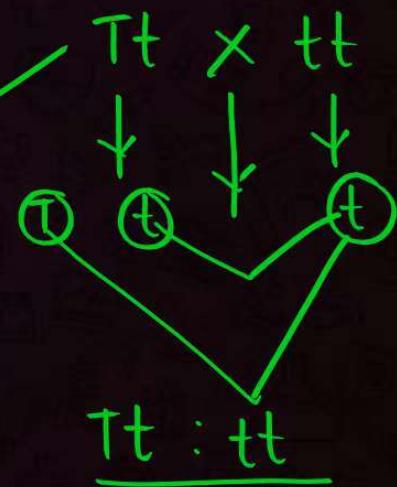
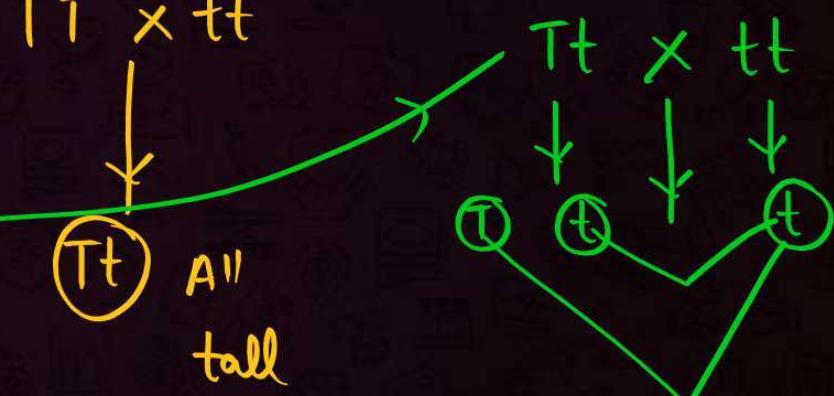
$Tt \times tt$

F₂ gen. \rightarrow F₂ gen \times Recessive
parent

Mendel
 F_2 gen. \rightarrow Tall
 (Used Test cross)

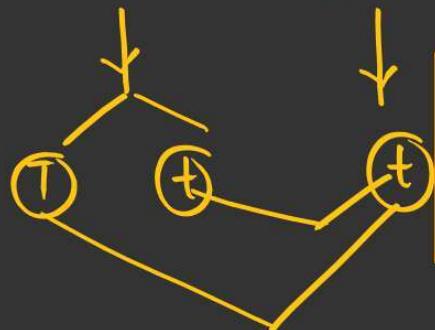


Test-cross \rightarrow utility used to check homozgosity / heterozygosity of the individual showing dominant phenotype



Monohybrid Test Cross

$$Tt \times tt$$



$$P.R = G.R = 1:1$$

Genotypes = Phenotypes

$$Tt \rightarrow \text{Tall}$$

$$tt \rightarrow \text{dwarf}$$

Phenotypes \rightarrow 2

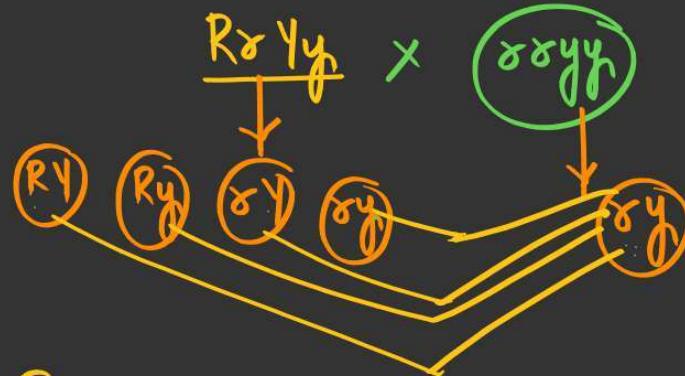
Genotypes \rightarrow 2

$$P.R = \text{Tall : dwarf} \\ 1 : 1$$

$$G.R = Tt : tt \\ 1 : 1$$

Dihybrid Test Cross

$$RrYy \times rryy$$



$$RrYy \quad Rryy \quad rryy \quad rryy \rightarrow \text{Genotypes}$$

Round yellow Round green wrinkled yellow wrinkled green

Phenotypes = Genotypes = 4

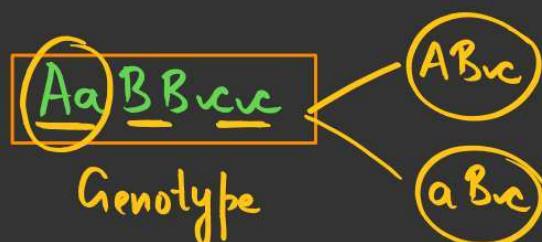
$$P.R = G.R = 1:1:1:1$$

Types of Gametes



$$2^n$$

n = No. of heterozygous conditions



$$(2^n) = 2^1 = 2$$

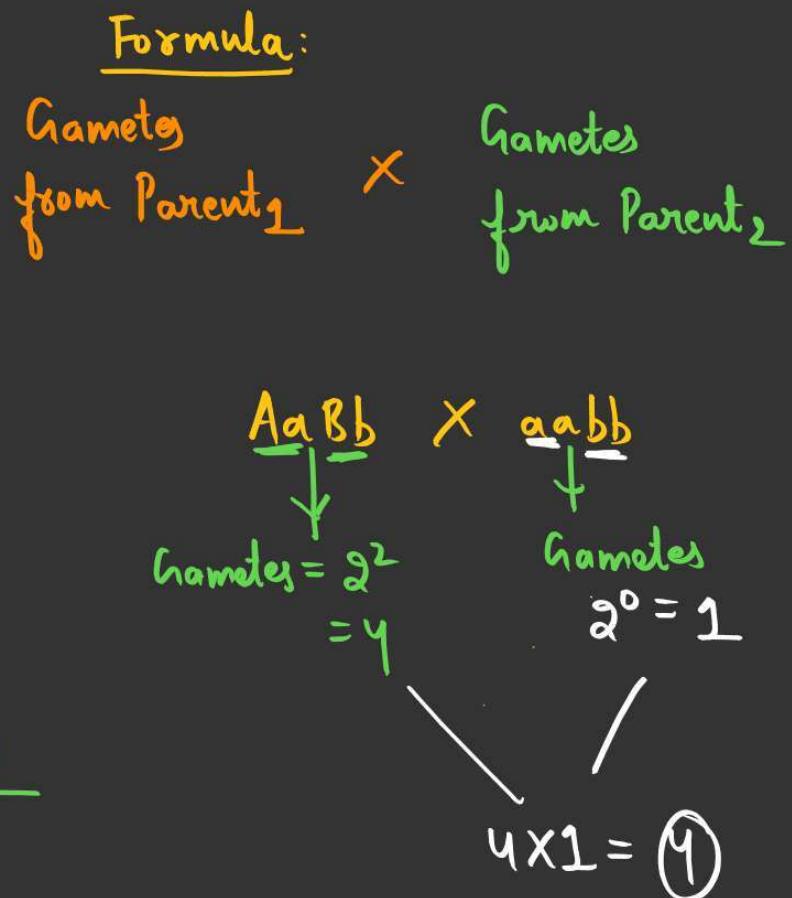
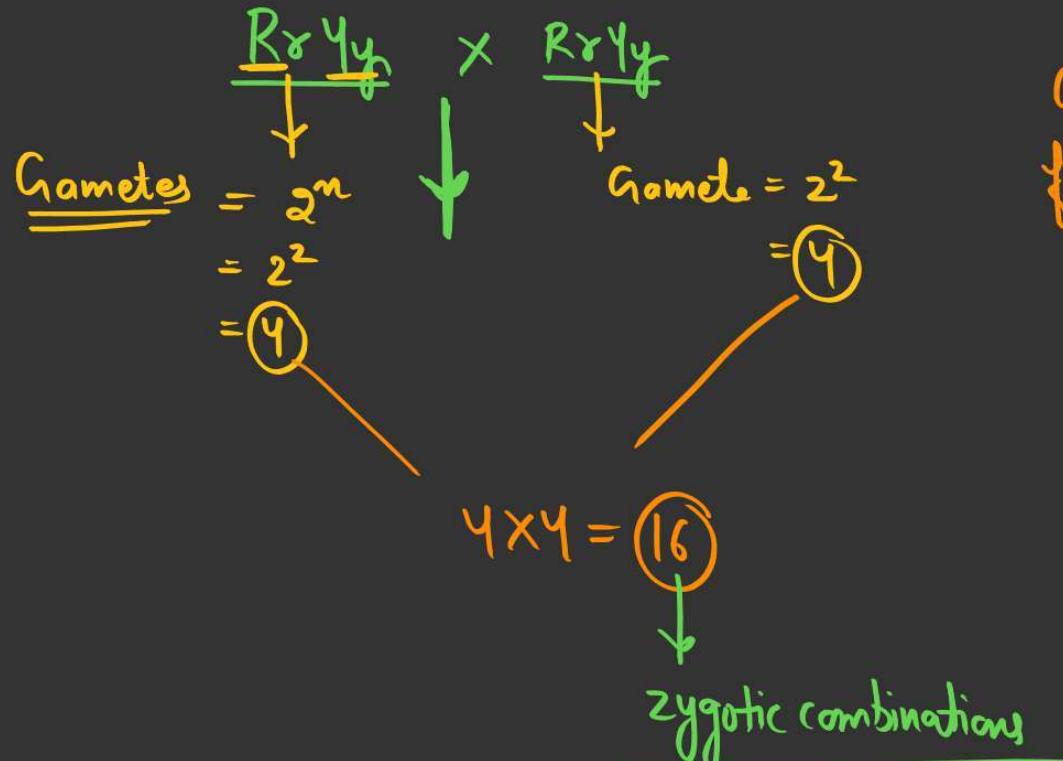
↳ An individual heterozygous for three different loci will form 3 different genes how many gametes?

Ans. $\boxed{Aa Bb Cc}$

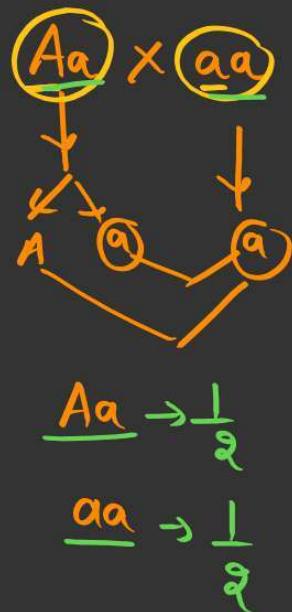
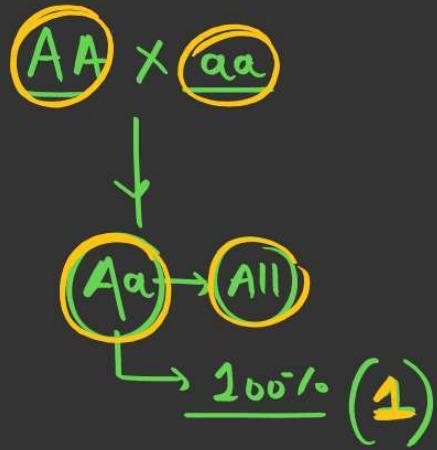
Ex $F_1 = \boxed{Rr Yy Tt}$
 $2^2 = 4$

$$(2^n) = 2^3 = 8$$

Number of offspring/zygotic combinations



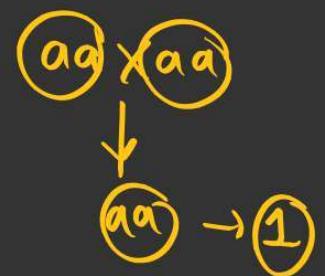
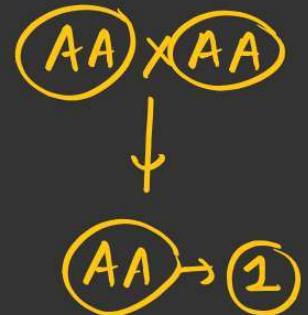
Frequency method



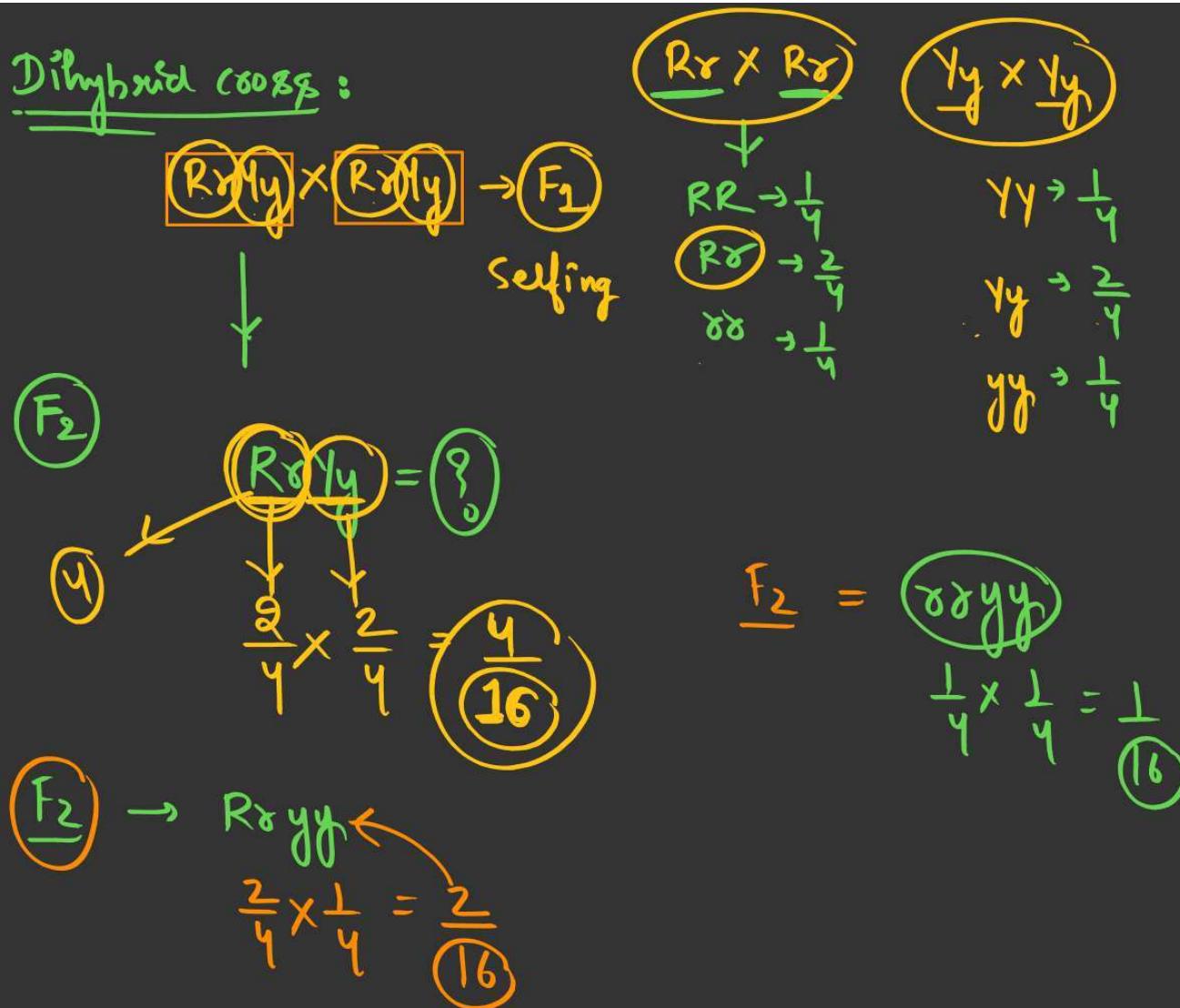
$$\begin{aligned} \text{Aa} &\rightarrow \frac{1}{2} \\ \text{AA} &\rightarrow \frac{1}{2} \end{aligned}$$

$$\begin{aligned} \text{Aa} \times \text{Aa} & \\ \text{AA} &\rightarrow \frac{1}{4} \\ \text{Aa} &\rightarrow \frac{2}{4} \\ \text{aa} &\rightarrow \frac{1}{4} \end{aligned}$$

A	AA	Aa
a	Aa	aa



Dihybrid cross:



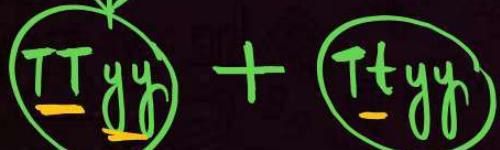
Q Cross between Tall Yellow ($TtYy$) and Tall green ($Ttyy$) is done. (Nice exercise)

Calculate the proportion of offsprings given below

$$(a) \text{ Tall and green} = \frac{3}{8}$$

$$(b) \text{ dwarf and green}$$

(a) Tall and green



$$\frac{1}{4} \times \frac{1}{2} + \frac{2}{4} \times \frac{1}{2} = \frac{3}{8}$$

$$Tt \times Tt$$

$$TT \rightarrow \frac{1}{4}$$

$$Tt \rightarrow \frac{2}{4}$$

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

$$Yy \times Yy$$

$$Yy \rightarrow \frac{1}{2}$$

$$yy \rightarrow \frac{1}{2}$$

$$(TtYy) \times (Ttyy)$$

$$\text{Gametes} = 4$$

$$\text{Gamete pairs} = 2$$

(b) dwarf & green

$$ttyy$$

$$\frac{1}{4} \times \frac{1}{2} = \frac{1}{8}$$



Q

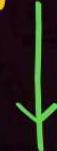
Heterozygous Round and yellow seeded plants were selfed.



800 seeds are collected. What is the total number of seeds

with first dominant and second recessive trait?

Ans (selfing) $\text{R}_y \text{Y}_y \times \text{R}_y \text{Y}_y$



$$\begin{array}{l}
 \text{R}_y \times \text{R}_y \leftarrow \begin{array}{l} \text{RR} \rightarrow \frac{1}{4} \\ \text{R}_y \rightarrow \frac{2}{4} \\ \text{YY} \rightarrow \frac{1}{4} \\ \text{Y}_y \rightarrow \frac{2}{4} \end{array} \\
 \text{Y}_y \times \text{Y}_y \leftarrow \begin{array}{l} \text{YY} \rightarrow \frac{1}{4} \\ \text{Y}_y \rightarrow \frac{2}{4} \end{array}
 \end{array}$$

$$\begin{array}{rcl}
 16 & \rightarrow & 3 \\
 1 & \rightarrow & \frac{3}{16}
 \end{array}$$

$$\begin{aligned}
 \text{Total seed} &= \frac{3}{16} \times 800 \\
 &= 150 \text{ seeds}
 \end{aligned}$$

F₂ gen.

offspring → Round green Y_y = $\frac{1}{4}$

seed

800

RRYY

$$\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$$

$\text{R}_y \text{Y}_y$

$$\frac{2}{4} \times \frac{1}{4} = \frac{2}{16}$$

$$\frac{1}{16} + \frac{2}{16} = \frac{3}{16}$$



Exceptions of Mendelism

- a) Incomplete dominance
- b) Complete dominance
- c) Multiple allelism.

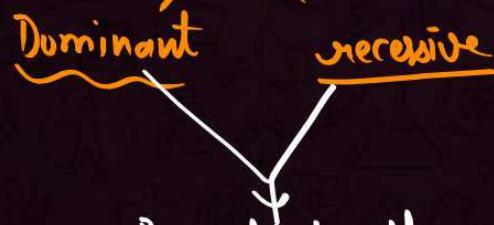
Incomplete dominance → support Blending inheritance



Ex → In flower color of Mirabilis jalapa (4 o'clock plant)

" " " " Snapdragon / Dogflower
(Antirrhinum majus)

* Two different form of a gene



Present together → Dominant allele show incomplete dominance

Dogflower

Red (RR) \times $\gamma\gamma$ (white)

↓

F₁ R γ (Pink)

R γ \times R γ \rightarrow Selfing

In F₂

Genotypes = ③

Phenotypes = ③

$$P \cdot R = \text{Red : Pink : white}$$
$$1 : 2 : 1$$

(F₂)

		R	γ
R	(Red)	(Pink)	
	RR	R γ	
γ	(Pink)	(white)	
	R γ	$\gamma\gamma$	

$$G \cdot R = RR : R\gamma : \gamma\gamma$$
$$1 : 2 : 1$$

$$G \cdot R = P \cdot R$$

Co-dominance

Two different forms of a same gene

→ Present together → Express equally



A-bld grp

B bld grp

Co-dominant alleles

AB, Blood group

+ Example of
Co-dominance

*

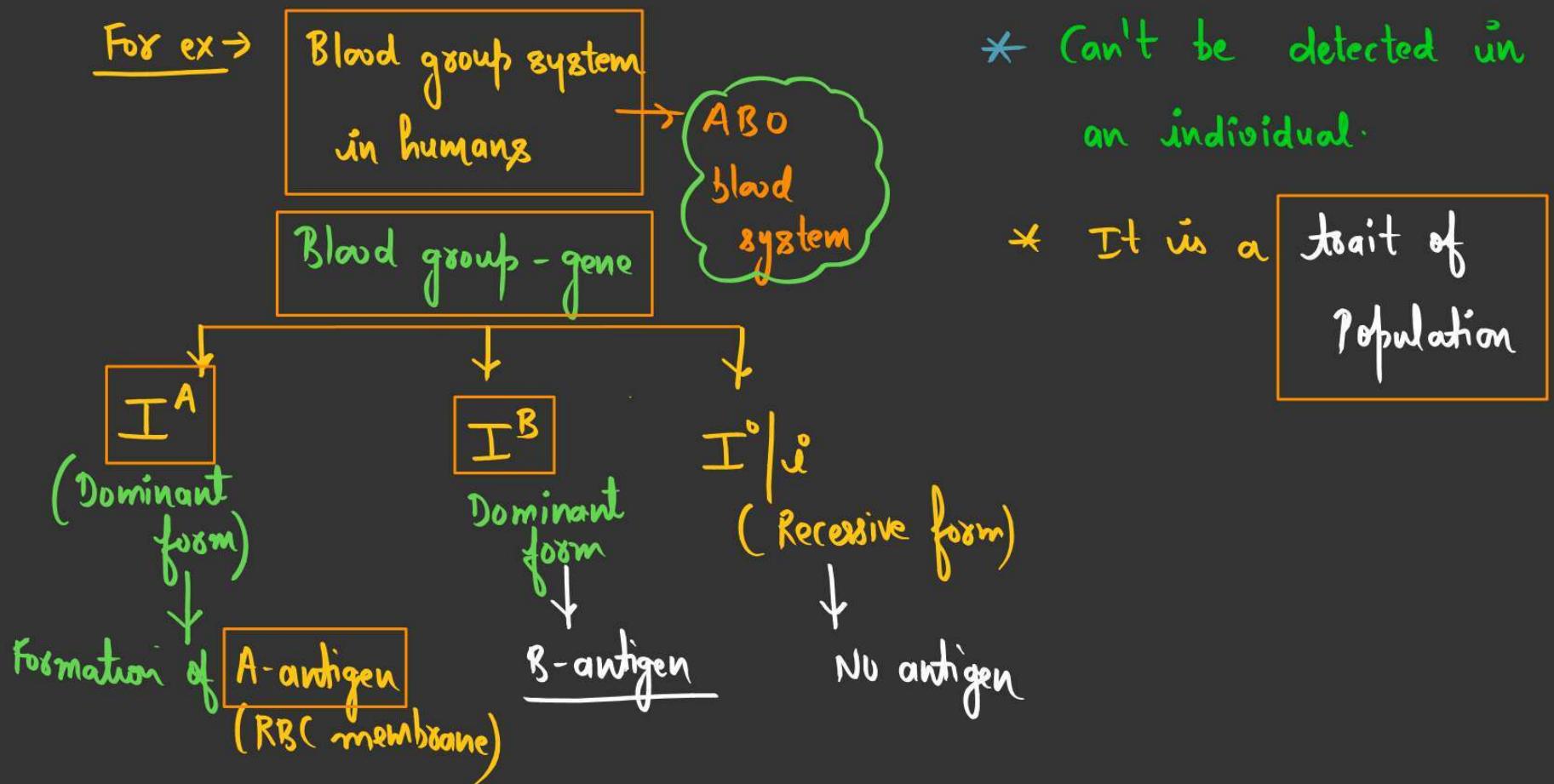
No of phenotypes = No. of genotypes

$$P.R = G.R = 1:2:1$$

Multiple allelism

When more than 2 alleles of a gene are present in a population.

For ex→



Genotype
 $I^A I^A$
 $I^A I^0$

Phenotype (Blood group)

(A)

$I^B I^B$
 $I^B I^0$

(B)

Population

Phenotypes \rightarrow 4

Genotypes \rightarrow 6

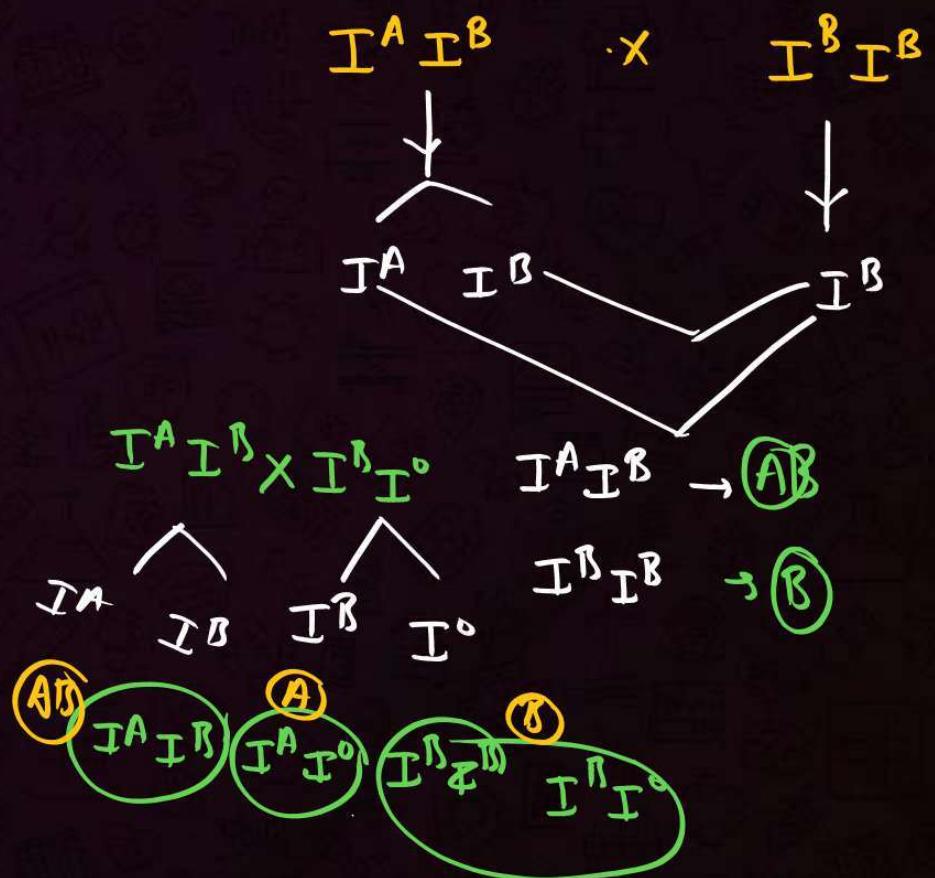
$I^A I^B \rightarrow AB$

$I^0 I^0 \mid \ddot{u} \rightarrow O$

Q What will be the possible blood group in children from the parents B and AB blood groups?



- a) A, O
- b) A, B, AB and O
- c) A, B, AB
- d) B, O



PLEIOTROPY

A pleiotropic gene affects / controls several phenotypes or traits

Example: a)

\underline{Hb}^S gene (Mutated gene) (causes Sickle anaemia)

b) Mutated gene causing disease Phenylketonuria

c) β -gene → Starch Branching enzyme gene

* Underlying mechanism: the product of this gene is related to many other pathways

B-gene → Pisum sativum

Controls ② Phenotypes

Starch grain
synthesis

Seed - shape

B-gene

Starch-grain
Size

Seed shape

Round

BB

Long

* when B-gene
controls starch-grain

Bb

Intermediate
(Incomplete
dominance)

Round
(Complete
dominance)

bb

Small

wrinkled

size
↓
Shows

Incomplete
dominance



Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.





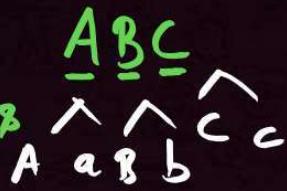
POLYGENIC INHERITANCE

- * When a character is controlled by many genes (Polygenes)
(≥ 3 or more than 3 genes)
- * Polygenic trait is not present in two distinct forms
but they are present across a gradient
- * Quantitative inheritance
The expression is affected by
number of dominant alleles
- * Effect of each dominant allele is additive

Ex → Skin color in Humans

→ Height in Humans

→ Intelligence in Humans



Skin-color → controlled
③ genes
(Polygenes)



AABBCC
⑥ dominant alleles
(Very dark) (Negro)

aabbcc
Total alleles → ⑥
(0 dominant allele)
very light (Albino)



AaBbCc
(3 dominant allele) → Intermediate
(Mulatto)



Rediscovery of Mendel's work

In 1900

Hugo de Vries

Carl Correns

Tschermak

* 1901 → "Republished"

Reasons why Mendel's work was not recognized?

- a) Communication at that was not easy.
- b) His methods of Mathematical & Statistical tools was first in field of Biology → which was not acceptable by other scientists.
- c) He could not provide "physical proof" of existence of factors.
(Microscopy was not developed)
- d) His concept of factors → "discrete units"
↓
do not blend
Was not accept by his contemporaries (Darwin)

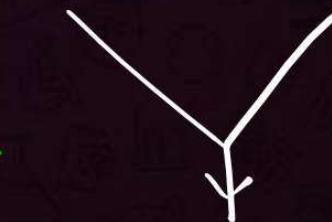


Chromosomal theory of inheritance

↓ By Sutton and Boveri → "1902"

- ↓
 - * Electron microscopy was developed.
 - * Chromosomes can be seen moving during division.

Mendel's work was Rediscovered



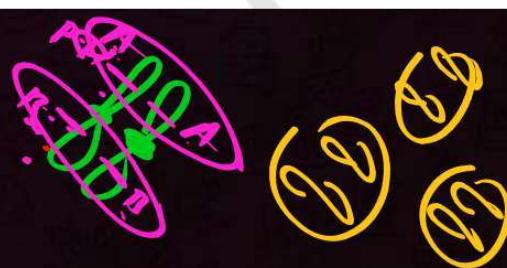
United the Knowledge of Mendel's work and Electron microscopy studies

Table 4.3: A Comparison between the Behaviour of Chromosomes and Genes



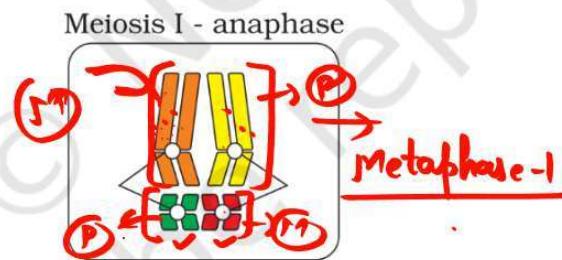
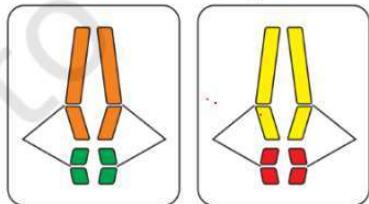
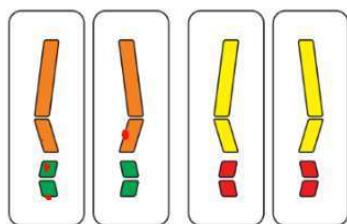
A (Gene)	B (Chromosome)
Occur in pairs <i>in diploid organism</i>	Occur in pairs
<u>Segregate</u> at the time of gamete formation such that only <u>one of each pair</u> is transmitted to a <u>gamete</u>	<u>Segregate</u> at gamete formation and only one of each pair is transmitted to a gamete
<u>"Independent pairs"</u> segregate independently of each other	One pair segregates independently of another pair → <i>Not always right for gene</i>

Can you tell which of these columns A or B represent the chromosome and which represents the gene? How did you decide?



Possibility I

One long orange and short green chromosome and long yellow and short red chromosome at the same pole


Meiosis II - anaphase - II

Germ cells

Possibility II

One long orange and short red chromosome and long yellow and short green chromosome at the same pole

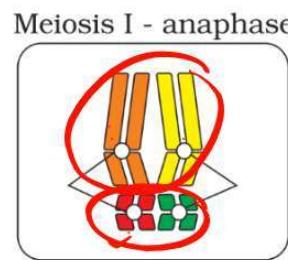
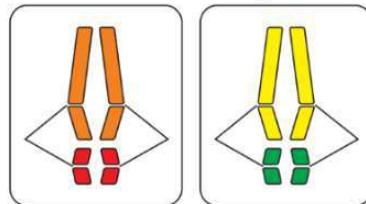
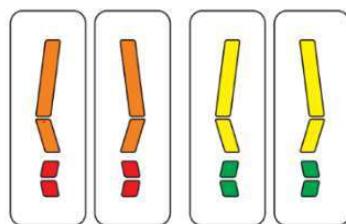
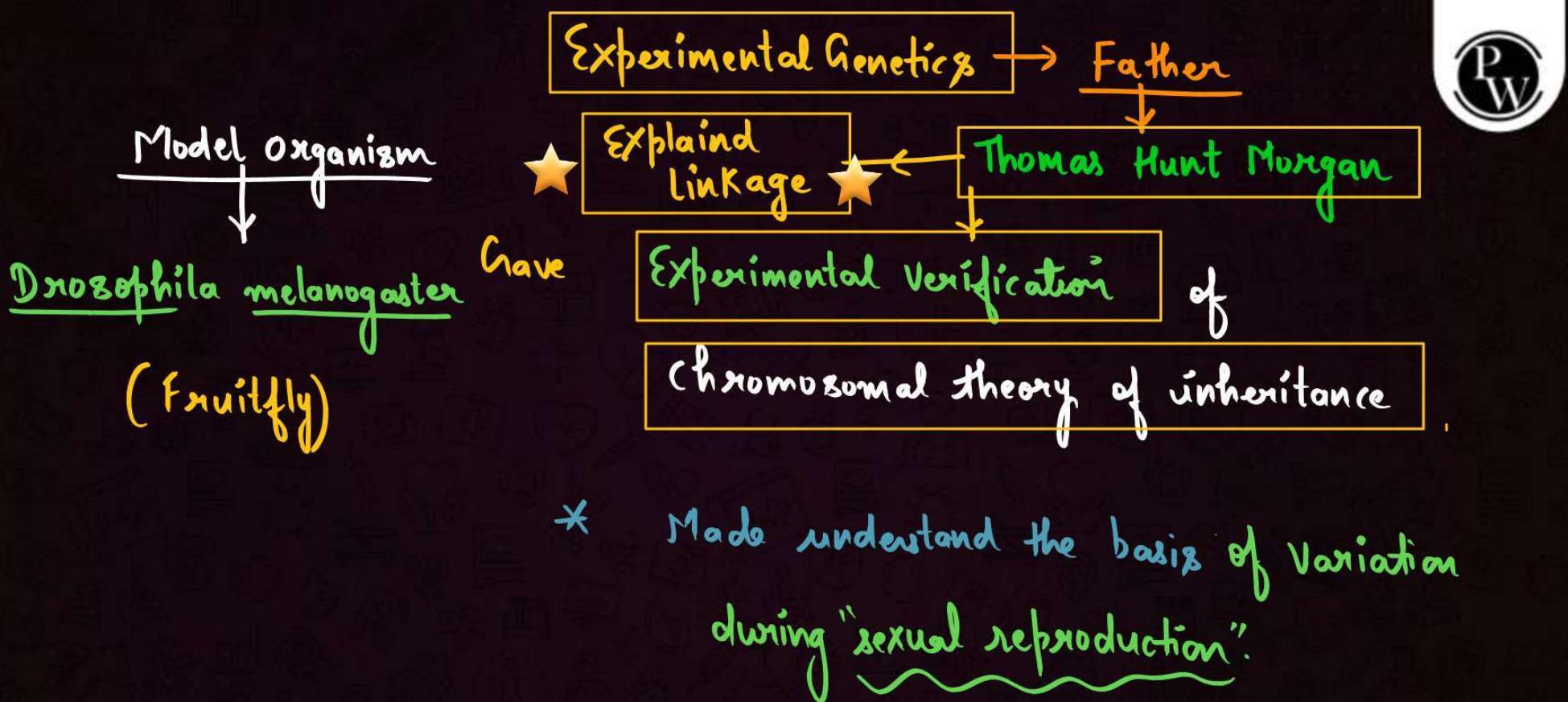

Meiosis II - anaphase

Germ cells


Figure 4.9 Independent assortment of chromosomes



* Made understand the basis of Variation
during "sexual reproduction".

Why Morgan chose Drosophila

1. life-span short (2-weeks)
2. Produce large number of offspring in single mating.
3. Can be easily grown in Synthetic medium
4. Male & females are easily distinguishable.
 ↓
 Longer.

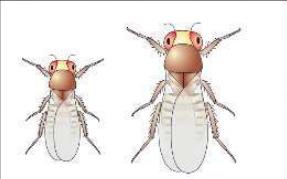


Figure 4.10 *Drosophila melanogaster* (a) Male
 (b) Female

5. Males have Heteromorphic sex-chromosomes.

Females



Males



6. Heredity Variations can
be studied in "low power
microscope".

Drosophila

Diploid organism

$2n = 8$ → ④ pairs of
chromosomes



↓
Males

6 + XY
(3 pair)
(Autosomes)

Allosomes
(sex chromosomes)

↓
6 + XX

LINKAGE



→ All the genes present on same chromosome
are physically associated & are
inherited together

This phenomenon is called as

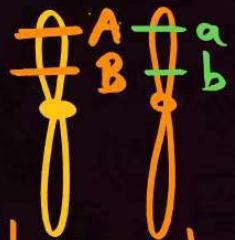
linkage.

P
W

Linkage

Complete linkage

AaBb



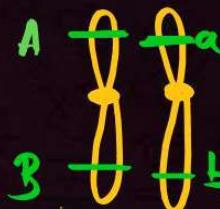
"No crossing over"

Gametes



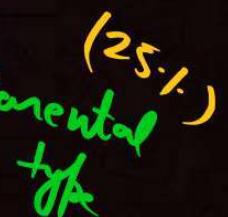
- * No recombinants
- * 100% Parental type

Incomplete linkage



→ Crossing-over can occur and can form recombinants

↓ ④ gametes



more
P.S.

Parental type

25% Recombinants

25% Parental type

D allele
 t

R allele
 r

3 ways

T^+
 t^+
 t

Morgan Experiment \rightarrow "Sex-linked inheritance"

Drosophila

Inheritance of genes present on sex-chromosome

P W

"X-linked inheritance" (X-linked genes) (Present only on X-chromosome)

Eye-color gene

Dominant form

Red
(w^+)

Recessive

white
(w)

Body-color gene

D

Brown
(y^+)

R

Yellow
(y)

Wing-type gene

D

Normal

m^+

R
Miniature
(m)

...

P
W

Experiment-1 Eye-color gene & Body color gene

Female (Recessive)
(Yellow bodied & white-eyed)

$$\times \begin{array}{c} y \\ \hline w \\ w \end{array}$$

$$\downarrow \text{Gamete}$$

$$\times \begin{array}{c} y \\ \hline w \\ w \end{array}$$

Male (Dominant) (wild type)
(Brown bodied & Red-eyed)

$$\times \begin{array}{c} y^+ \\ \hline w^+ \\ w^+ \end{array}$$

$$\times$$

$$Y$$

Gamete

F₁ (Mating)
generation

(24188-18038
* inheritance)
(X-linked traits)

(Females)

$$\times \begin{array}{c} y^+ \\ \hline w^- \\ y^+ w^+ \end{array}$$

$$\times$$

$$\times \begin{array}{c} y \\ \hline w \\ w \end{array}$$

(Males)

(Brown & Red)

$$x yw$$

$$(y^+ w^+) \times$$

$$y^+ w$$

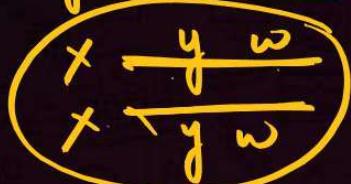
$$y^+ w$$

Yellow & white

$$yw \quad Y$$

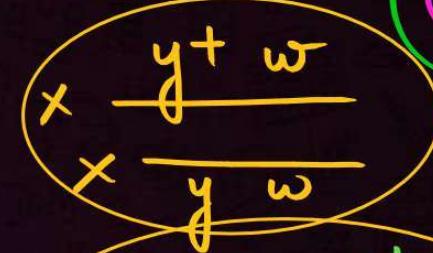
P
W

F₂ gen: Parental

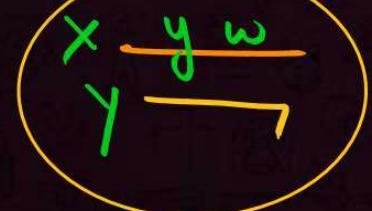
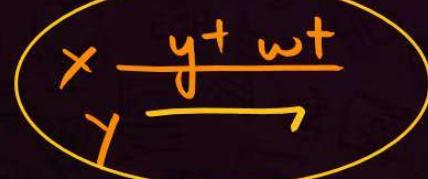
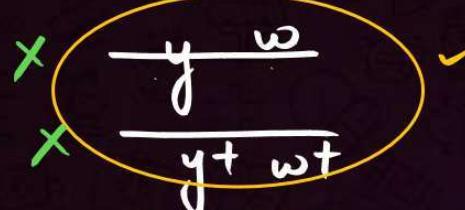


(98.7%)

Recombinants



(1.3%)





Female

Experiment - 2

$\frac{w\ m}{w\ m} \times \frac{w^+ m^+}{w^+ m^+}$

Eye-color gene \times wing type gene

"Same-cross"

F_2 gen

Result

\downarrow

Parental type

\downarrow

(62.8%)

Recombinants

(37.2%)

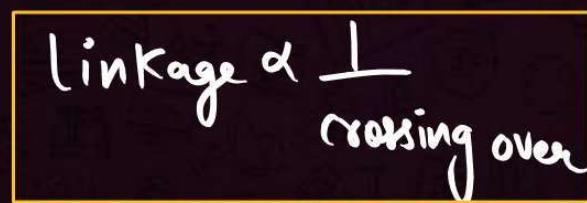
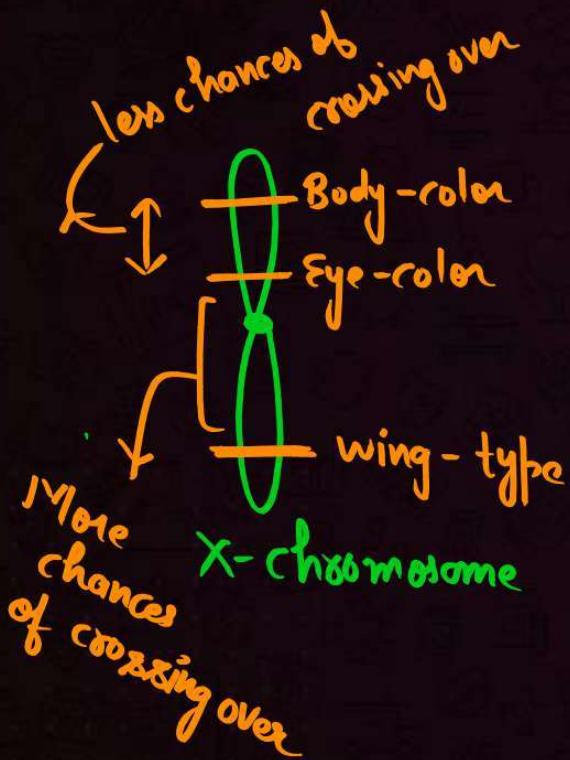
Conclusion



Distance between genes
on same chromosome



Crossing over | Recombination



Chromosomal mapping / Gene-mapping

↓ Alfred Struttevant (*Drosophila*)

- * Genes are arranged linearly on chromosome.
- * The arrangement of genes / Position of genes can be predicted

by **Rerombination frequency | Cross-over value
(C.O.V)**

is called as **chromosomal mapping**.



**R.F | C.O.V = Distance b/w
genes on a chromosome**



Distance b/w genes

**Centimorgan
(CM)
(Euk)**

**Map Unit
(M.U)
(Pso)**

Morgan's Experiment

Eye-color × Body-color

$$\underline{R.F} / \underline{C.O.V} = (1.3.4)$$

* Distance between
eye-color & Body color
gene on X-chromosome
is (1.3 cM)

Eye-color - wing type

$$R.F / C.O.V = (37.2\%)$$

$$\text{Distance} = (37.2 \text{ cM})$$



\therefore

$$\text{Distance between } A-B = 3 \text{ M.U}$$

$$B-C = 7 \text{ M.U}$$



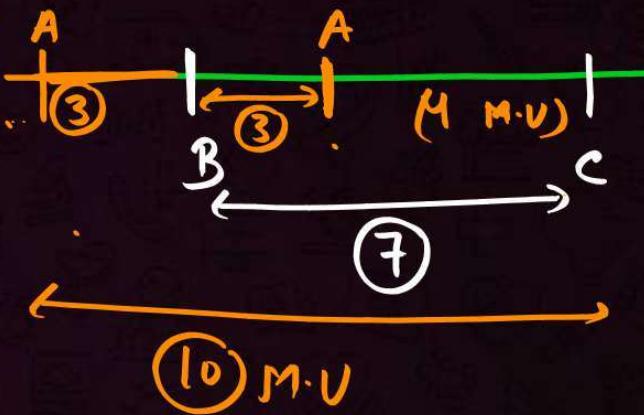
$$A-C = ?$$

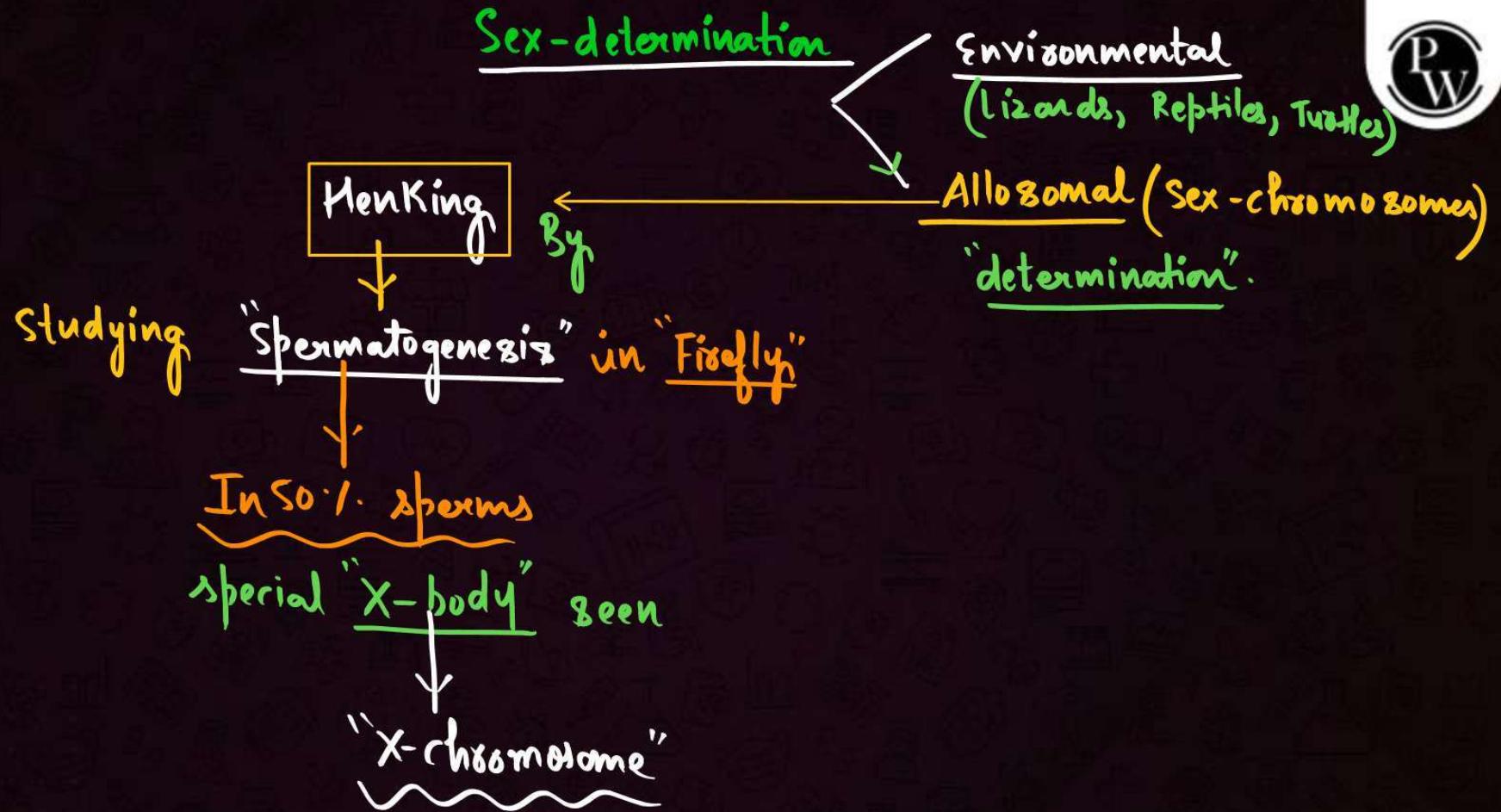
a) 4 M.U

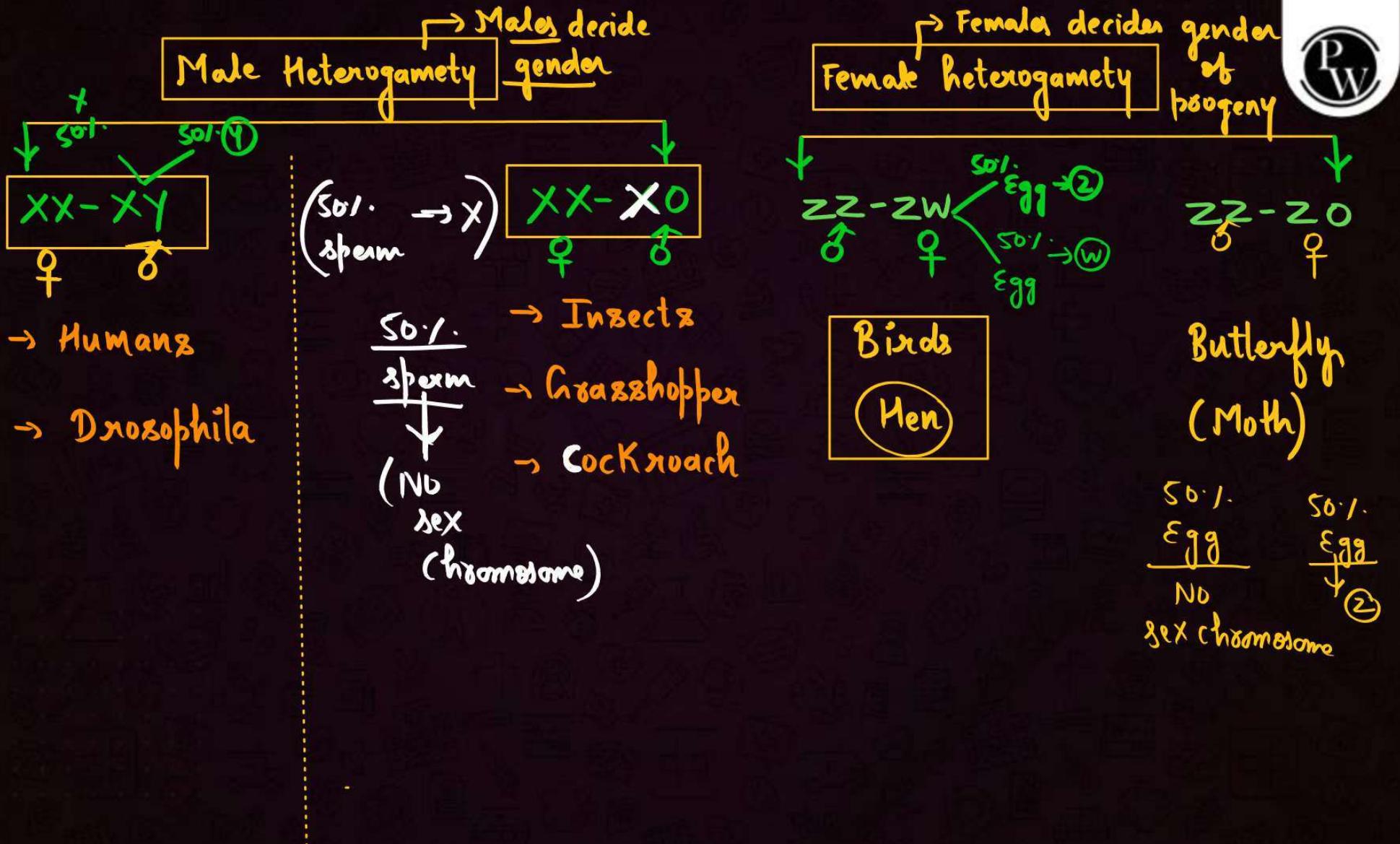
b) 10 M.U

c) 6 M.U

~~d)~~ Either a or b







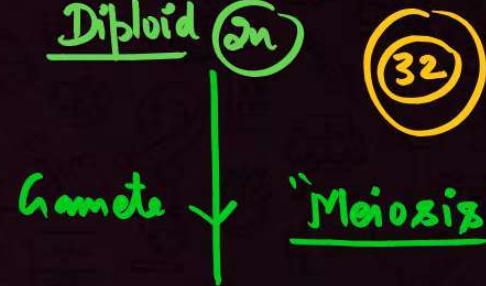
Honey-bee → Haplo-diploidy / Parthenogenesis

Males / Drones → Haploid



Females (Queen)

Diploid (2n)

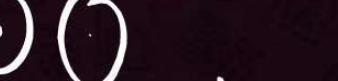


Male
gamete
(n)

(16) chromosomes

Female
gamete
(n)

(Fertilized) Male
gamete



female
gamete

(n) (2n) (Female)

(16) chromosomes

without
fertilization

Males
(Drone) (n)



Male

(Figure 4.13), they do not have father and thus cannot have sons, but have a grandfather and can have grandsons.

Due to
Mutagens

Mutation

"Sudden change in genetic
make-up" of an organism



↓
Gene-mutation
change occurs
in the sequence
of gene

↓
Genomic mutation
Change in number of
chromosomes

↓
chromosomal mutation/
chromosomal aberration
↓
Change in structure of
chromosome



Point mutation

change occurs
in single base
pair.

Ex → Sickle cell
Anaemia

Gene-mutation

base pair mutation

↓
change occurs
in many
base pairs.

Substitution

Transition



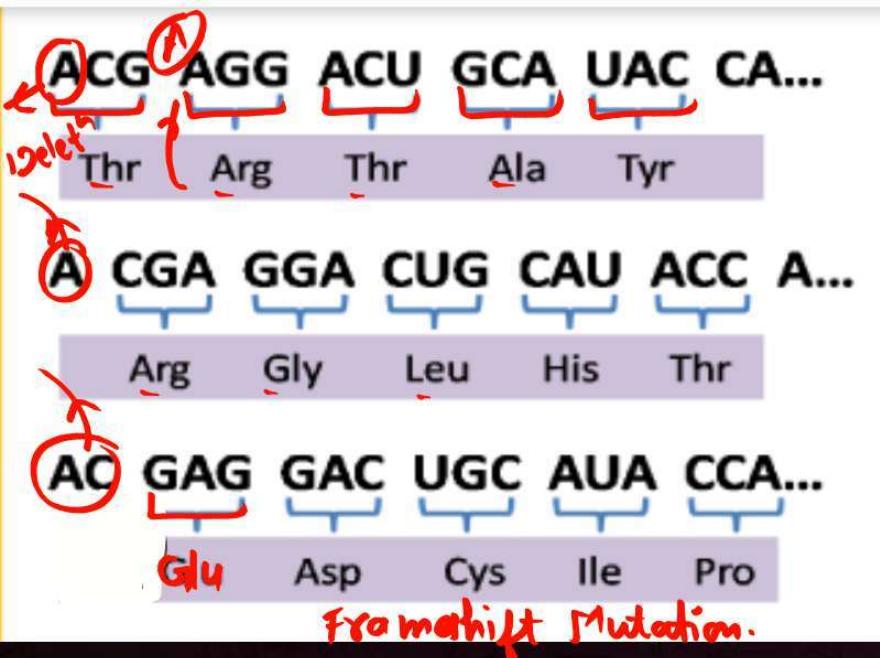
Purine replaced by Purine
Pyrimidine " " Pyrimidine

Transversion



Purine is replaced
by pyrimidine
(Vice-versa)

(Sickle-cell
Anaemia)



Addition | Deletion
↓
Frame shift Mutation → Thalassemia

P W

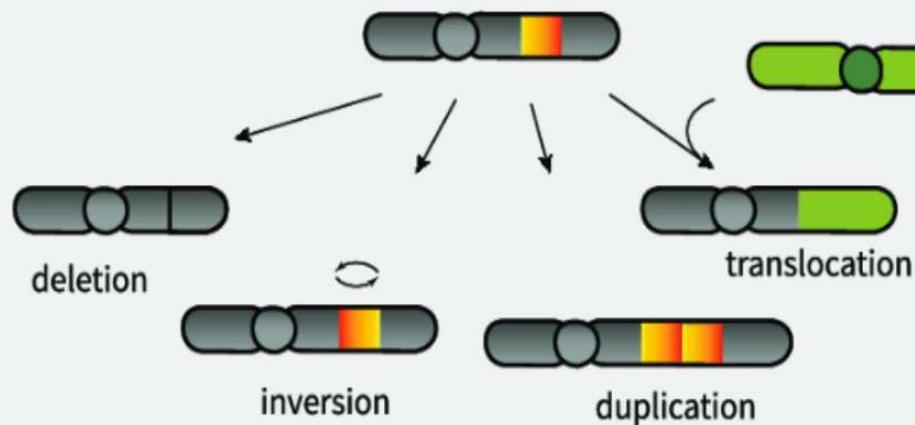
Addition Deletion
Insertion
Base / Bases
are added

Base / Bases → delete

A T G C T T T

A T G C ~~T T T~~
 (A)

b simple structural variations



Chromosomal mutations



Genomic mutation

P
W

Aneuploidy

[change do not occur in complete set of chromosomes]

but only in a pair or two]

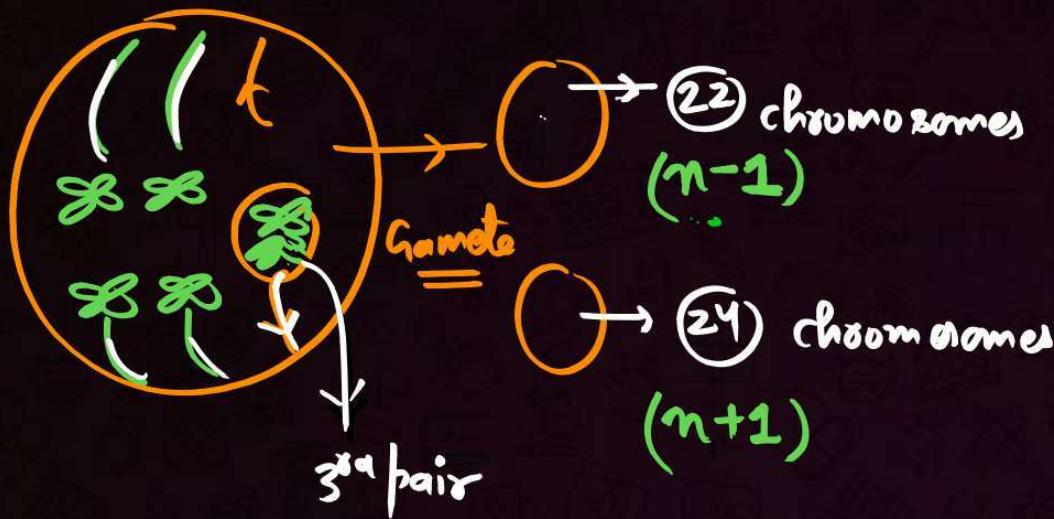
Reason: Non-disjunction of chromosomes during Anaphase
do not segregate / separate

Polyplody

* when complete set of chromosomes change

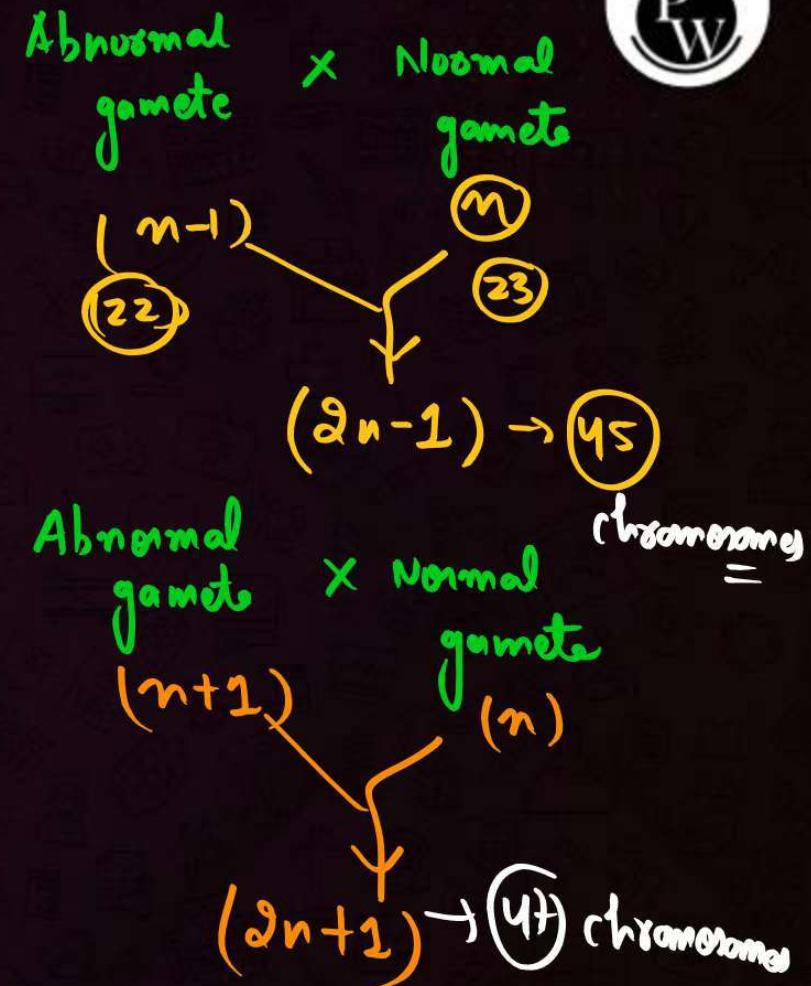
$$2n \rightarrow 4n$$

Reason: cell which is arrested at Anaphase failure of proper division



Gametes $n = 23$

Mitochondria



Aneuploidy

↓ Hyperploidy
(Increase)

Trisomy

↓
 $(2n+1)$ → 47 chromosomes

→ Down Syndrome

→ Klinefelter syndrome

↓ Hypoploidy
(Decrease)

$(2n-1)$ → 45 chr.

↓
MONOSOMY

EX → Turner Syndrome

Genetic disorders

P
W

Non-Mendelian disorders

Cause < Chromosomal mutation
Genomic mutation

Ex

- Down Syndrome
- Klinefelter syndrome
- Turner Syndrome

Mendelian disorders

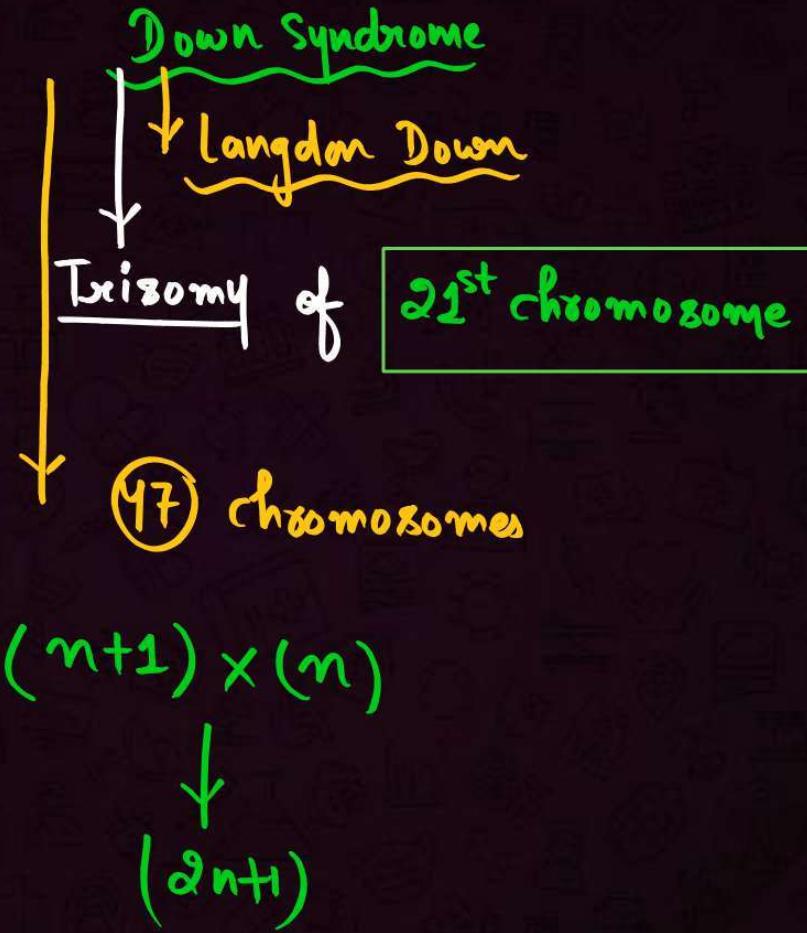
Cause → Gene mutation

↓
Autosomal

↓
Dominant
"Myotonic
Muscular
Dystrophy"

↓
Recessive
→ Sickle-Cell Anaemia
→ Thalassaemia
→ Phenylketonuria

↓
X-linked
"Recessive"
→ Colour
Blindness
→ Haemophilia



Symptoms:

- Moon-face
- Flat-head
- Partially open mouth
- Furrowed tongue
- Middle crease in palm
- Physical, psychomotor, Mental Retardation
- Congenital heart disease
- Short stature

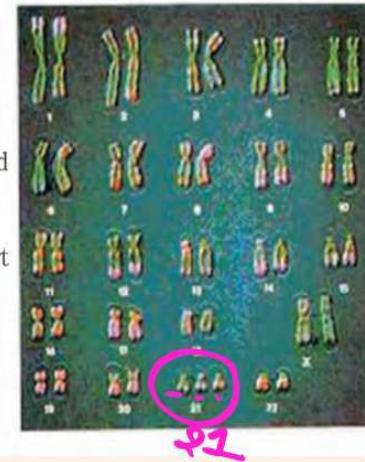
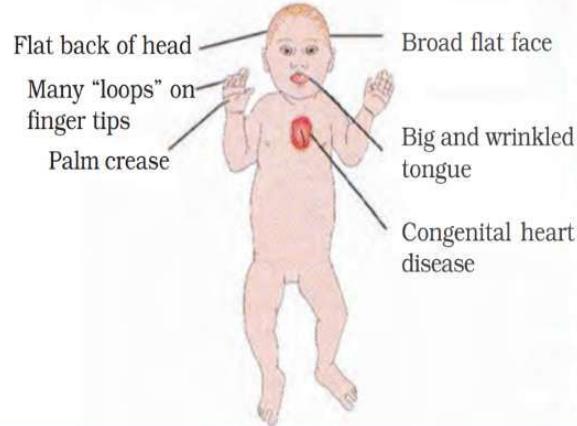


Figure 4.16 A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual

Normal male
 $44 + XY$

→ Trisomy of Klinefelter Syndrome
23rd pair (Sex-chromosomes)

→ In Males

→ $44 + XXY$

or

$47, XXY$

extra X-chromosome
in males.

Symptoms:

- Gynaecomastia
 ↓
 "Develop of Breasts"
 (Meminine characters)
- "Sterile"

Egg
Abnormal
 $22 + XX$

Normal sperm
 $22 + Y$

$(22 + X)$

$44 + XXY$

Normal X Abnormal
egg sperm
 $22 + XX$ $(22 + X)$
 ↓
 $44 + XXY$

Normal female
 $44+XX$

Turner Syndrome

Monosomy of 23rd pair \rightarrow (In females)

$44+X0$

or $45, X0$

(One X-chromosome)

Symptoms:

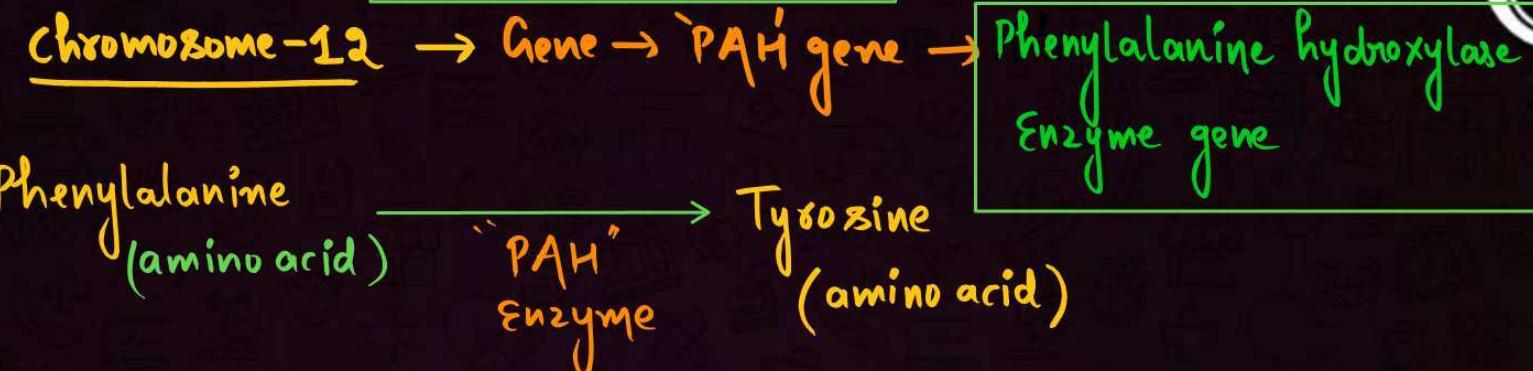
- sterile
- short stature
- Ovaries \rightarrow Rudimentary
- Secondary sexual characters not developed.

Abnormal gamete \times normal gamete
 $(n-1) \times (n)$

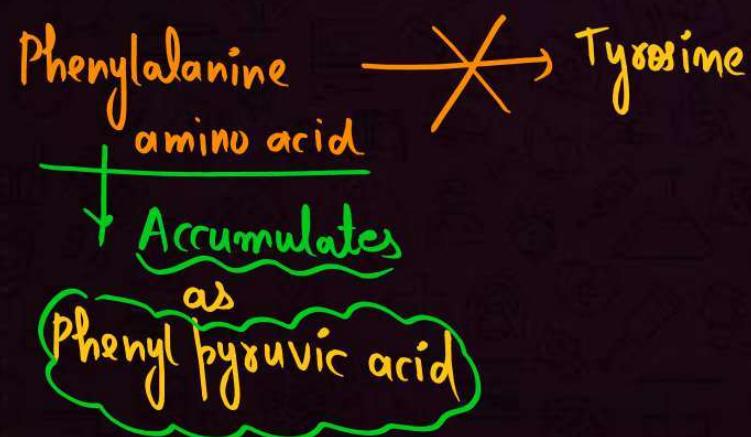
 $(2n-1)$



PHENYLKETONURIA



Chromosome - 12 → PAH Gene → Mutation → Becomes Recessive (will not form Enzyme)



Mutated Gene
(Pleiotropic gene)

Phenylpyruvic acid accumulates in Brain cells → Mental Retardation

↓
Excreted
in Urine

→ Depigmentation in
hair & skin



Thalassaemia → Needs Regular Blood Transfusion

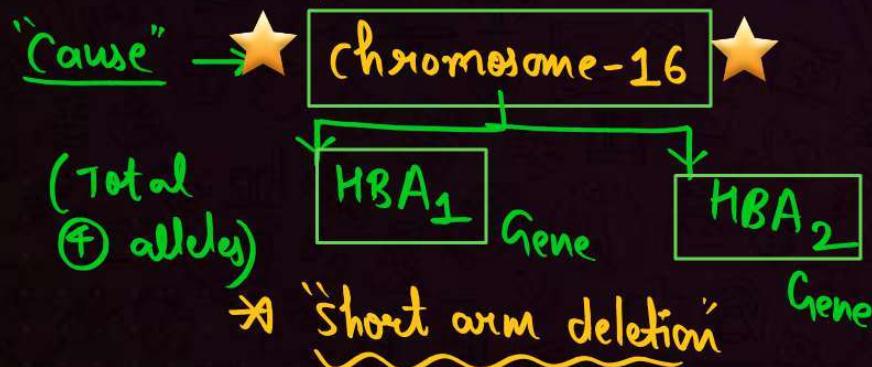


↓ α-Thalassaemia

* Only β-chain formed

* α-chain Not formed (Major)
β-chain formed (Minor)

* RBC → "Ruptures"



↓ β-Thalassaemia

β-chains Not formed
α-chain formed

Cause :

Chromosome-11

↓ HBB gene
(mutation)

Sickle-Cell Anaemia

- Point mutation
- Substitution (Transversion)
- Autosomal Recessive disorder

Hb^A gene
 ↓
Normal gene
 ↓
 Normal Haemoglobin

→ Hb^S gene → "Pleiotropic gene"
 (Mutated gene)
 (Abnormal Haemoglobin)

Hb^A Hb^A → Normal Hb^A Hb^S → Carrier Hb^S Hb^S → Diseased



Chromosome
11

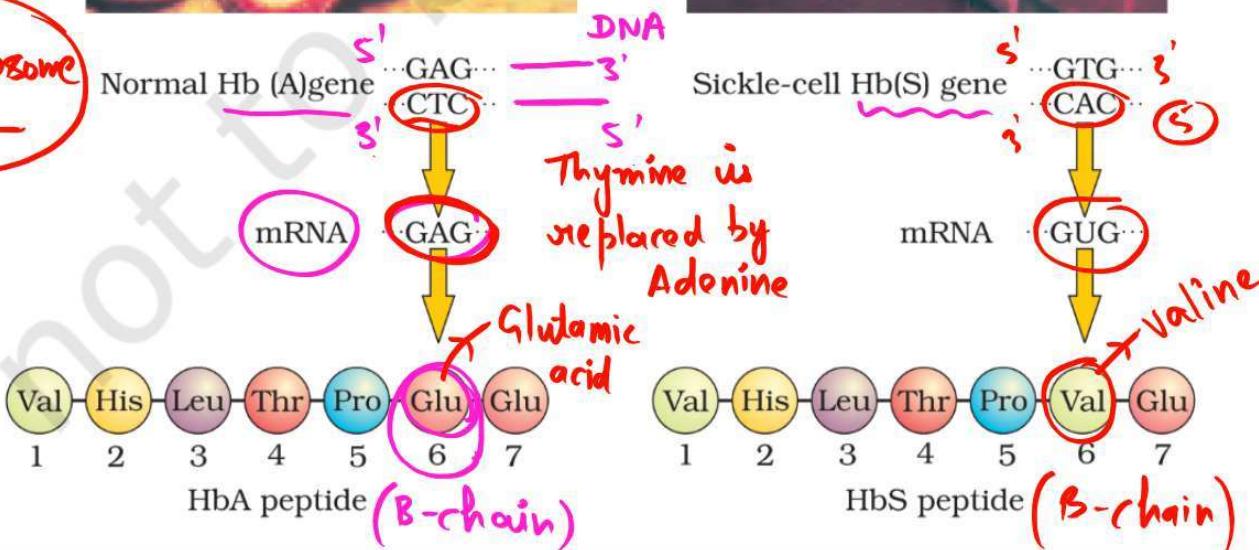


Figure 4.15 Micrograph of the red blood cells and the amino acid composition of the relevant portion of β -chain of haemoglobin: (a) From a normal individual; (b) From an individual with sickle-cell anaemia

Haemophilia

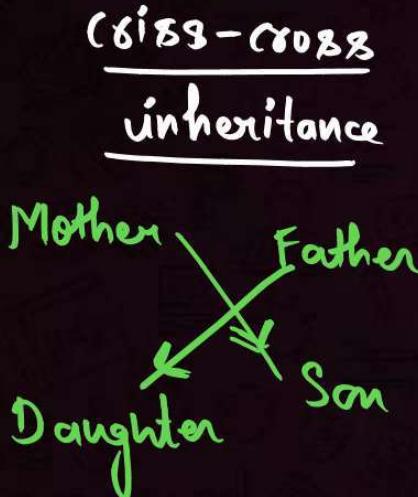
* X linked recessive disorder

* Bleeder's disease

* Coagulation factor (Any one of them)
 ↓
 (Cascade effect)

Blood coagulation is Absent

* "Royal disease" (Pedigree of Queen Victoria)



$XX \rightarrow$ Normal

$XY \rightarrow$ Normal

$XX^h \rightarrow$ Carrier

$X^h Y \rightarrow$ Haemophilic

$XX^h \rightarrow$ Haemophilic



Color-Blindness

\rightarrow X-linked recessive disorder

* Red-green color blindness

* Criss-cross inheritance

* Population  Males \rightarrow 8%

Females \rightarrow 0.4%

$XX \rightarrow$ Normal

$XY \rightarrow$ Normal

$XX^c \rightarrow$ Carrier

$X^c Y \rightarrow$ Color Blind

$XX^c \rightarrow$ color blind



X-linked recessive disorders

Color Blindness

Haemophilia

are more common in Males than in Females

Reason:

Females
↓

② recessive
copies must
be present

Males

① recessive

copy on X-chromosome
is enough for disease.

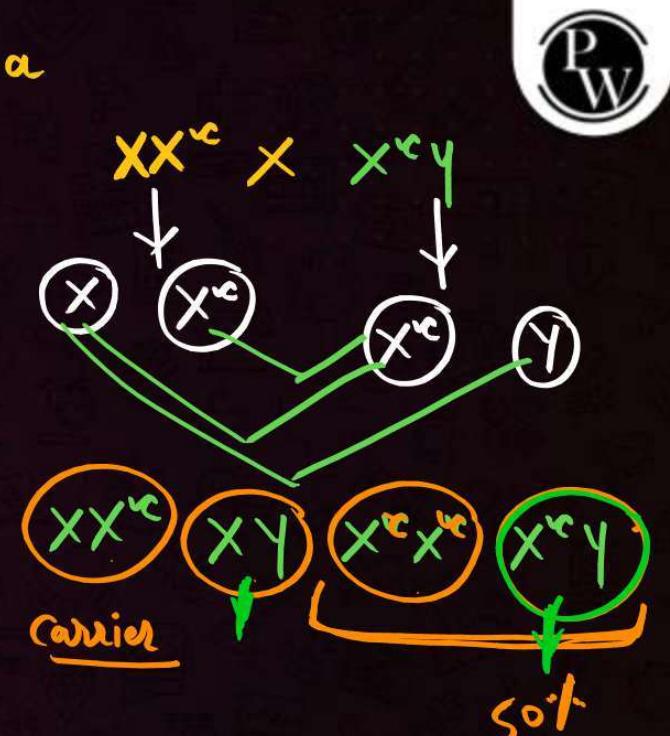


B A female whose father was color-blind marries a color-blind man. Calculate the following?

a) what % of their progeny will be diseased
 $\hookrightarrow 50\%$

b) what % of males will be color-blind
50%

c) what % of females will be color-blind
 $\downarrow 50\%$



Pedigree-chart

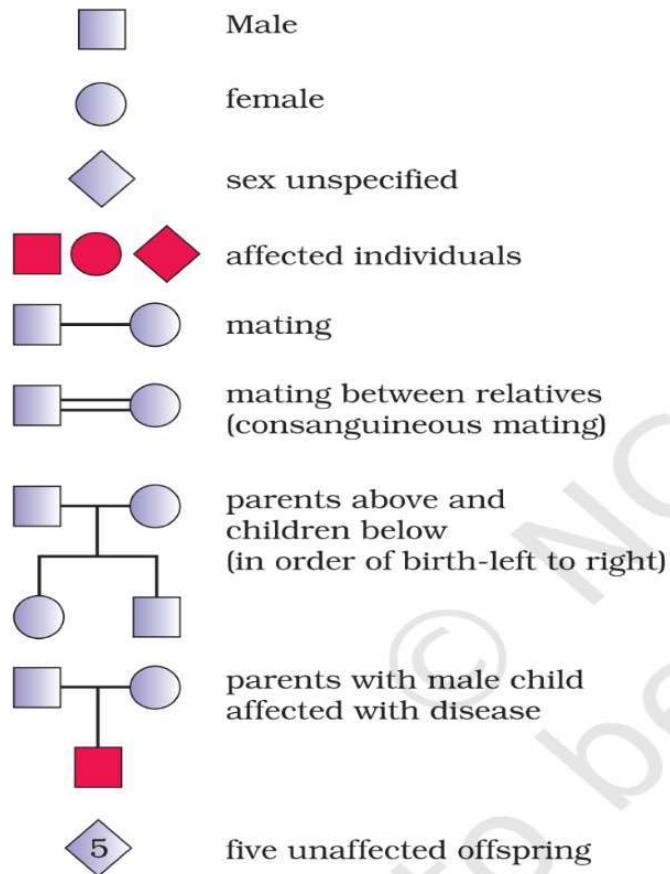
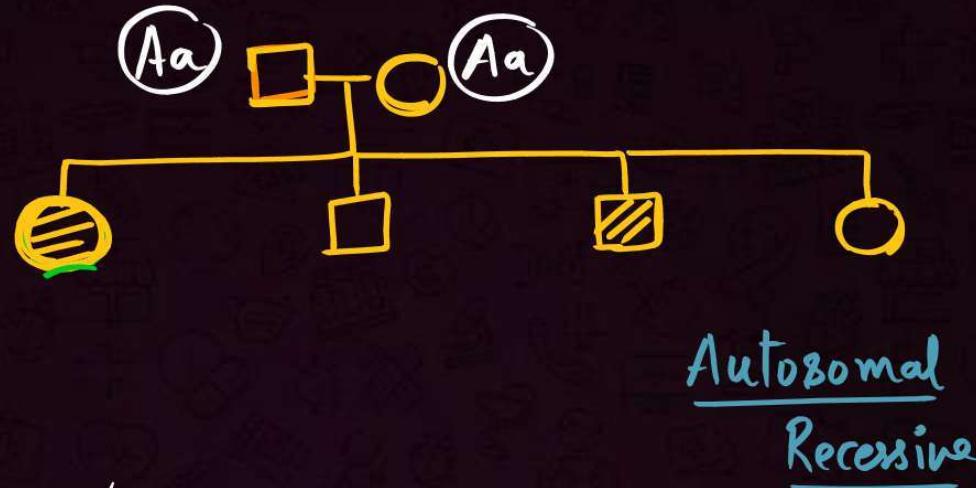


Figure 4.13 Symbols used in the human pedigree analysis

- ✓ Autosomal dominant
- ✓ " recessive
- ✓ X-linked dominant
- ✓ " recessive



$aa \rightarrow$ Diseased
 AA
 Aa
 } Normal

\oplus This chart is
Autosomal Recessive
 Predict genotype of parents

Fundae

① If parents are Normal



Progenies → Diseased

Cannot be

Dominant chart

X-linked
dominant

Autosomal
dominant

② Father → Normal
Daughter → Diseased

Cannot be

X-linked
Recessive chart

③

Mother → diseased
Son → Normal

→ Can't be
X-linked
Recessive
chart.

④ Mother → Normal
Son → Diseased

Cannot be X-linked Dominant chart

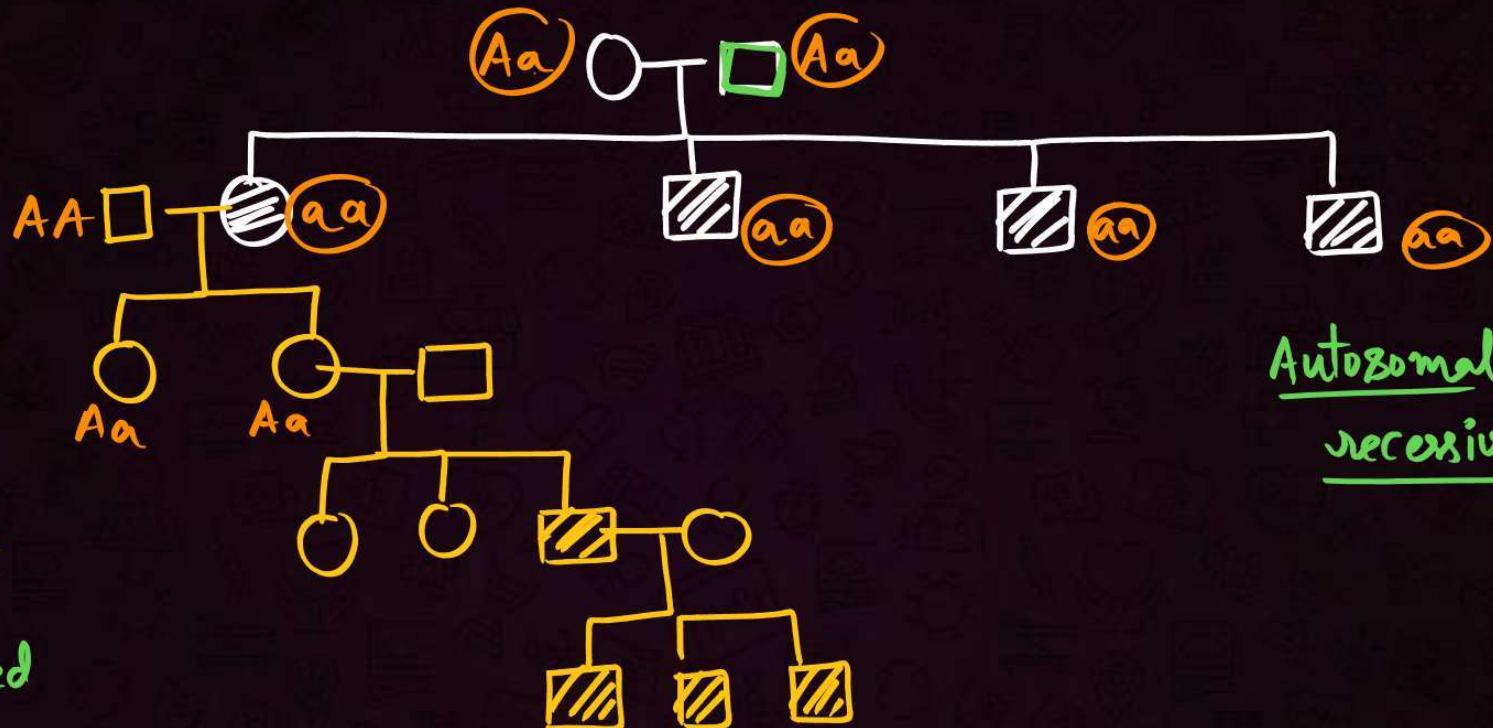
PW

PW

~~Autosomal dominant~~

~~X-linked dominant~~

~~X-linked recessive~~

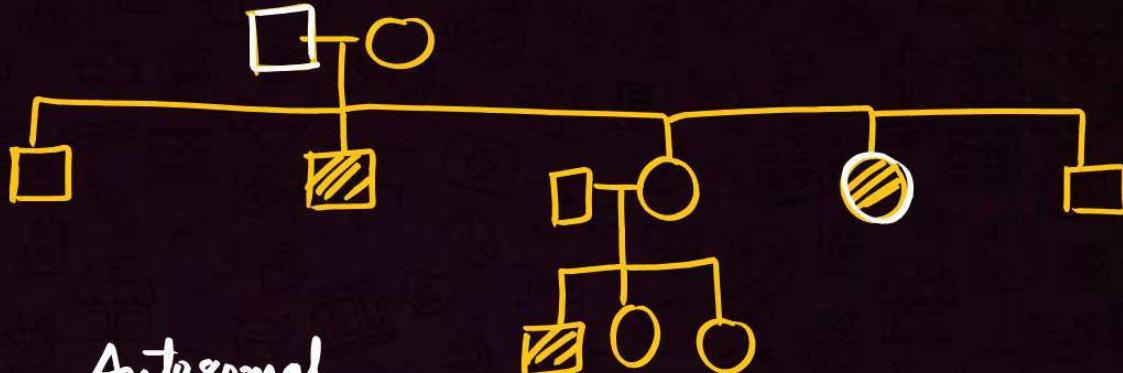


Autosomal recessive

P_W

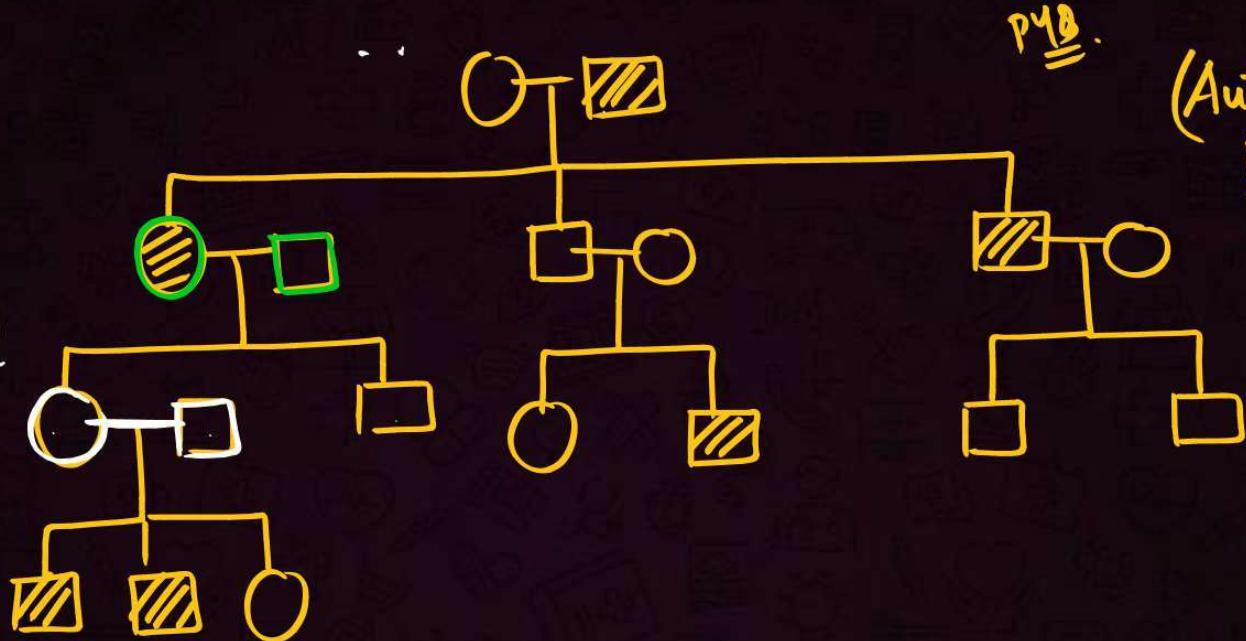
~~Dominant
chart~~

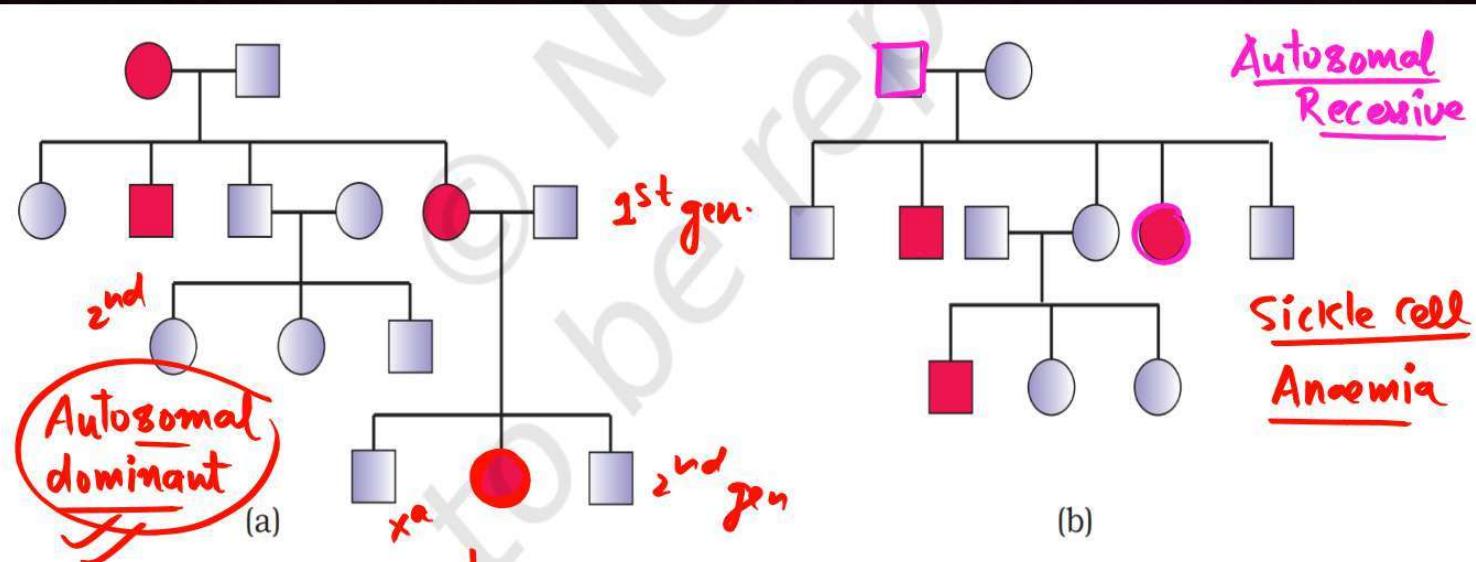
~~X-linked
recessive~~



Autosomal
recessive

~~Dominant~~
~~Chart~~
X-linked Autosomal
~~X-linked~~
~~recessive~~





* Myotonic Muscular Dystrophy *

