PRACHAND NEET

ONE SHOT

Botany

Principles of inheritance and Variation

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Genetics → Branch of Science

Inheritance

Process of transmission of characters from one generation to another.

* **Heredity**

Phenomenon of inheritance of genes

Variation

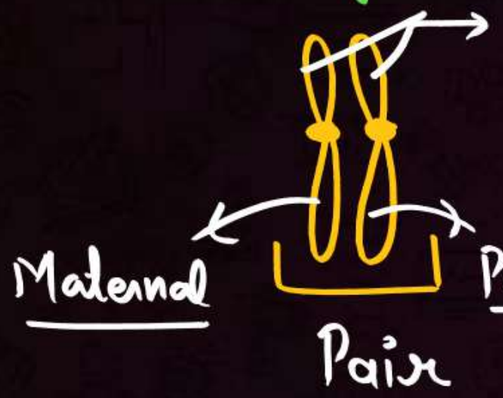
Degree of difference between parents & progeny/ offsprings.

Cause

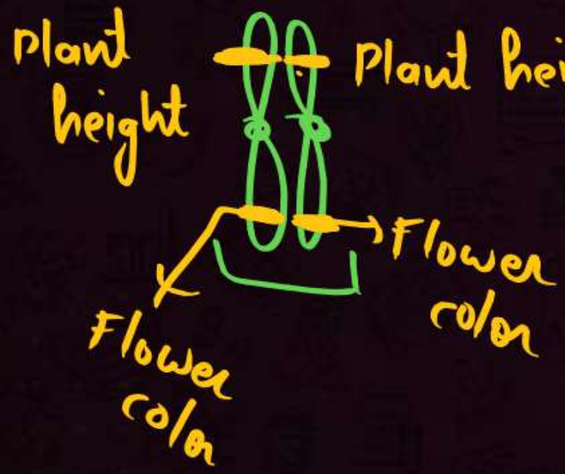
- Recombination
- Mutation (Major)

Terminology

Homologous chromosomes



Size, structure & types of genes are similar



In diploid organism (In diploid cell)

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

Genes are in pair (Each gene has (2) copies)

NOTE: In haploid organism / In haploid cell

Each chromosomes has single copy

Genes has single copy

Concept of Dominance

DNA

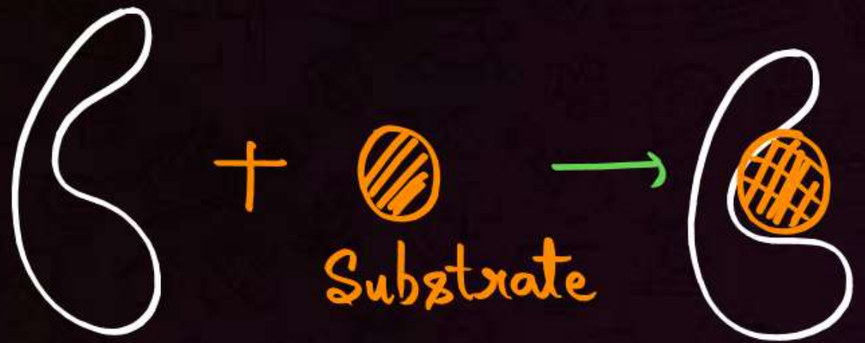
"Gene" → Codes for information

↓ Transcription

m-RNA

↓ Translation

→ Protein (Act as Enzyme)



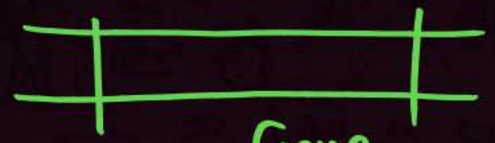
Enzyme

↓

Responsible for Transformation of Substrate

Product

(Responsible for phenotype expression).



Gene

Original gene / Wild gene / Normal gene / Unmodified gene / Dominant gene

Mutation

Modified allele / Mutated allele

↓

"Recessive allele"

Forms

a) No enzyme at all

b) Forms "non-functional enzyme"

* Mostly the mutated alleles are Recessive alleles

Modified allele

Equivalent to Unmodified allele

↓

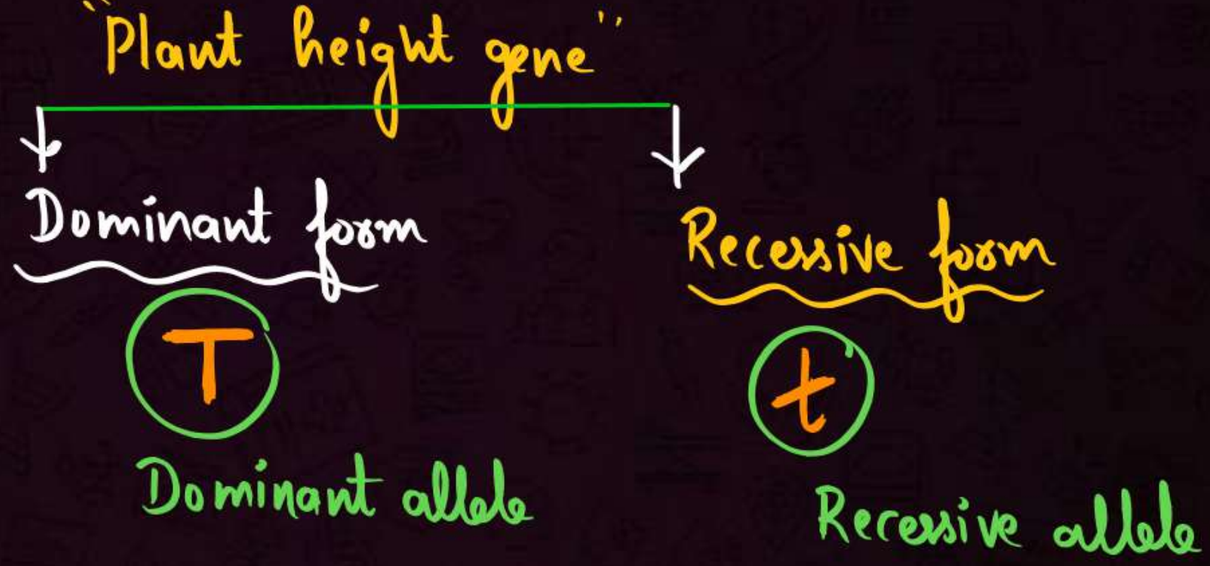
Such equivalent pairs are very common.

a) Forms Same Enzyme

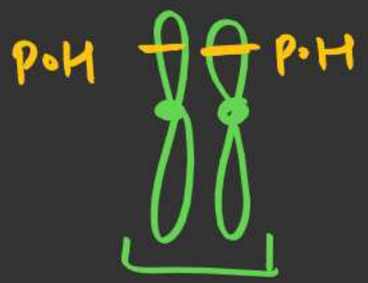
b) Forms less efficient enzyme (same enzyme)

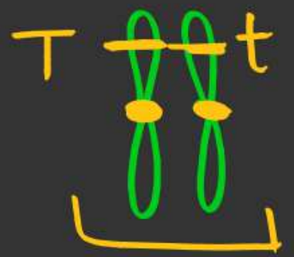
Alleles / Allelomorphic pair

Different forms of a same gene.



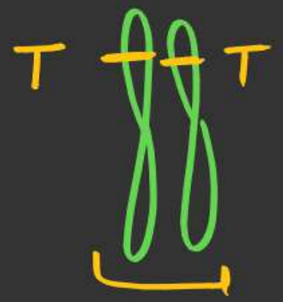
Heterozygous



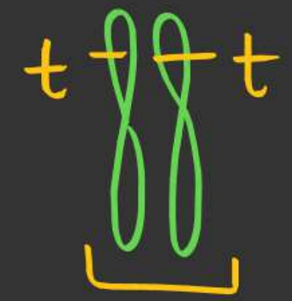


Tt

Homozygous



TT



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**

Feauture of an organism

* Flower color
* Seed shape

Trait

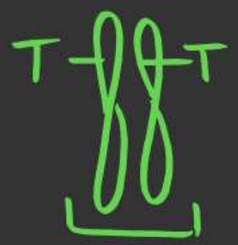
Distinguishable form of character

Plant height (character)
- Tall → Trait
- Dwarf → Trait

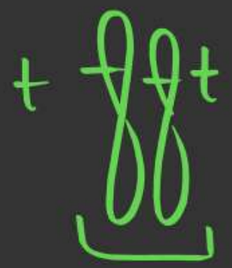
Seed shape
- Round → Trait
- wrinkled → Trait

Pure line / True-breeding variety

Homozygous for a **character** / Trait



(TT) Pure line



(tt) Pure line

Ncert 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

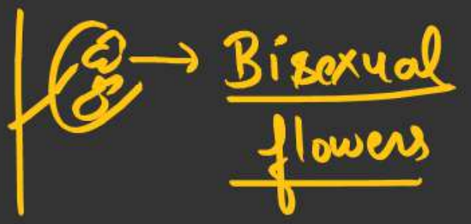
* worked on Pisum sativum (Pea plant)

for 7 years (1856-1863) PhD.

* work was published in 1865.

Pisum sativum (why)

a) Short life span



→ Bisexual flowers

b) Produce large no. of offsprings / progeny (Seeds)

c) Easy to grow in lab.

d) Has seven pair of Contrasting traits (imp)

e) Naturally self-Pollination.

f) Cross-pollination by Artificial hybridisation.

Pisum sativum

Diploid organism

$2n = 14$

7 pairs of chromosomes.

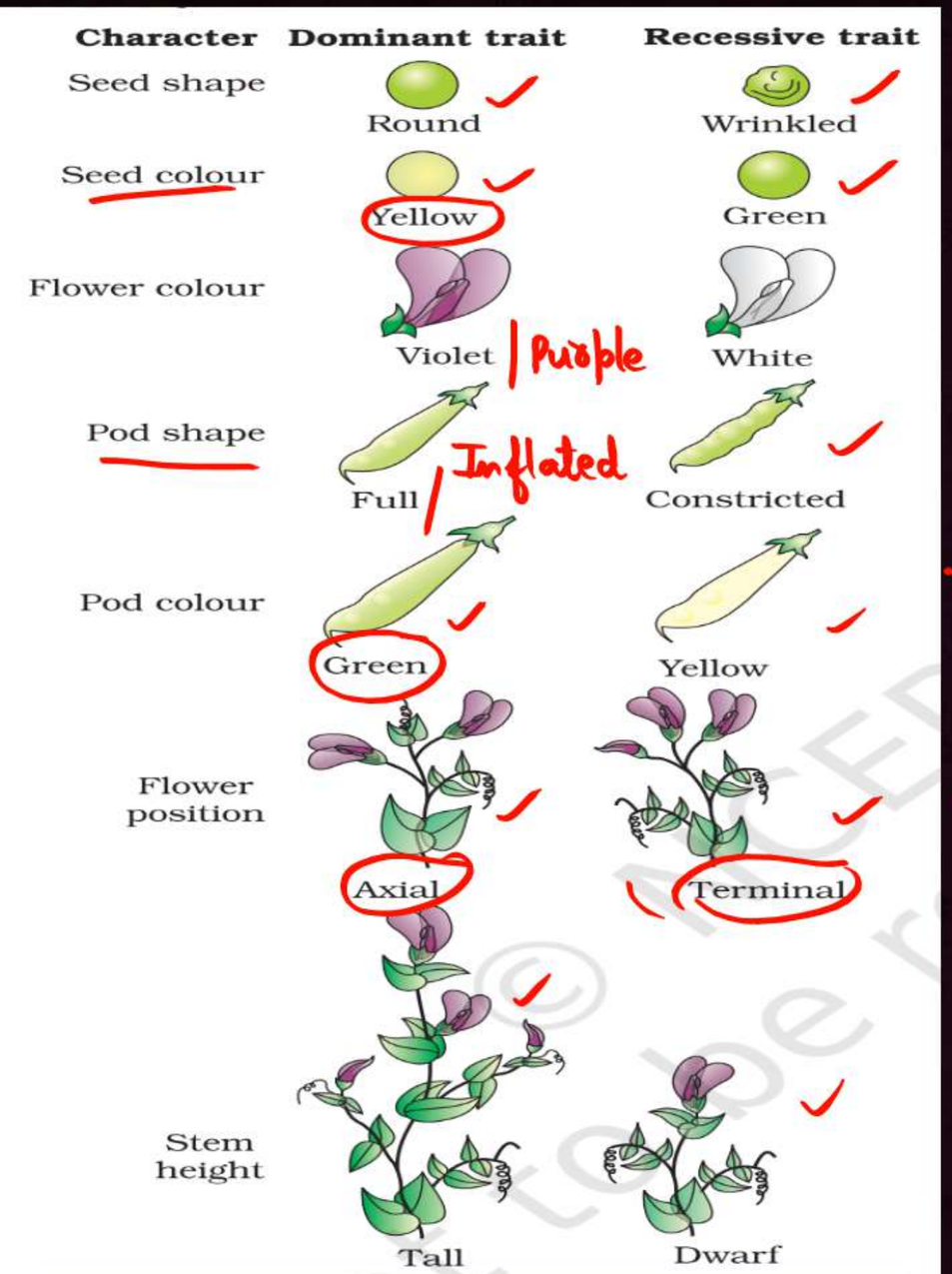
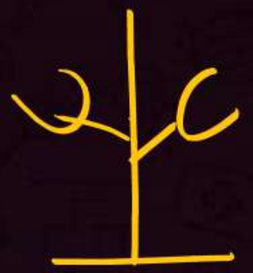
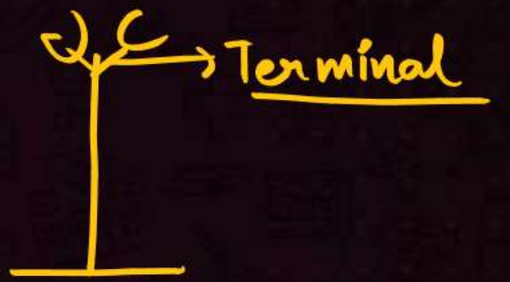


Figure 4.1 Seven pairs of contrasting traits in pea plant studied by Mendel



Axial
(sidewise)



Trait → Dominant

trait → Recessive

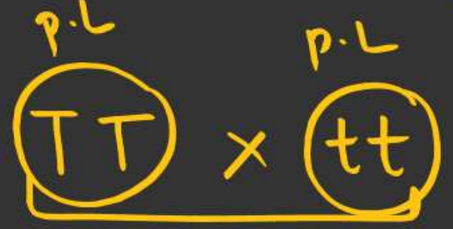
Mendel was Successful

① Mathematical & statistics tools in Biology
(was the first)

② He took **large sample size**

Mendel took

(14) true-breeding variety | Pure lines



Experiment-1

Exp. -(2)

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.

Monohybrid cross

Parents: True breeding / **Pure line**

Cross pollination / Artificial hybridisation

TT (Tall) × tt (dwarf)

T

Gametes: t

(F1 Hybrids) Tt → F1 generation / (All Tall) First filial generation

Phenotype is similar to Dominant parent.

Tt × Tt **Selfing** (Self-pollination)

T t Gametes

♂ / ♀	T	t
T	TT Tall	Tt Tall
t	Tt Tall	tt dwarf

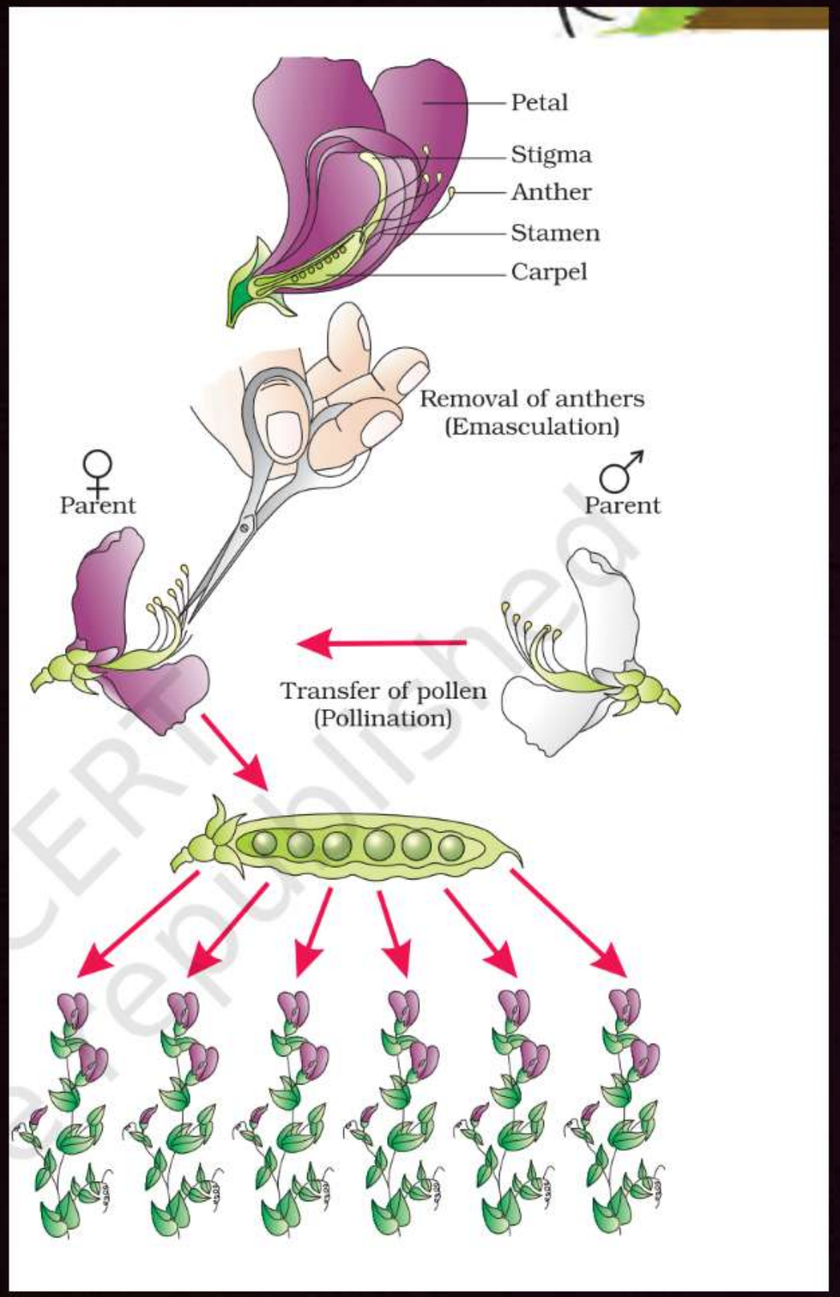
F2 generation

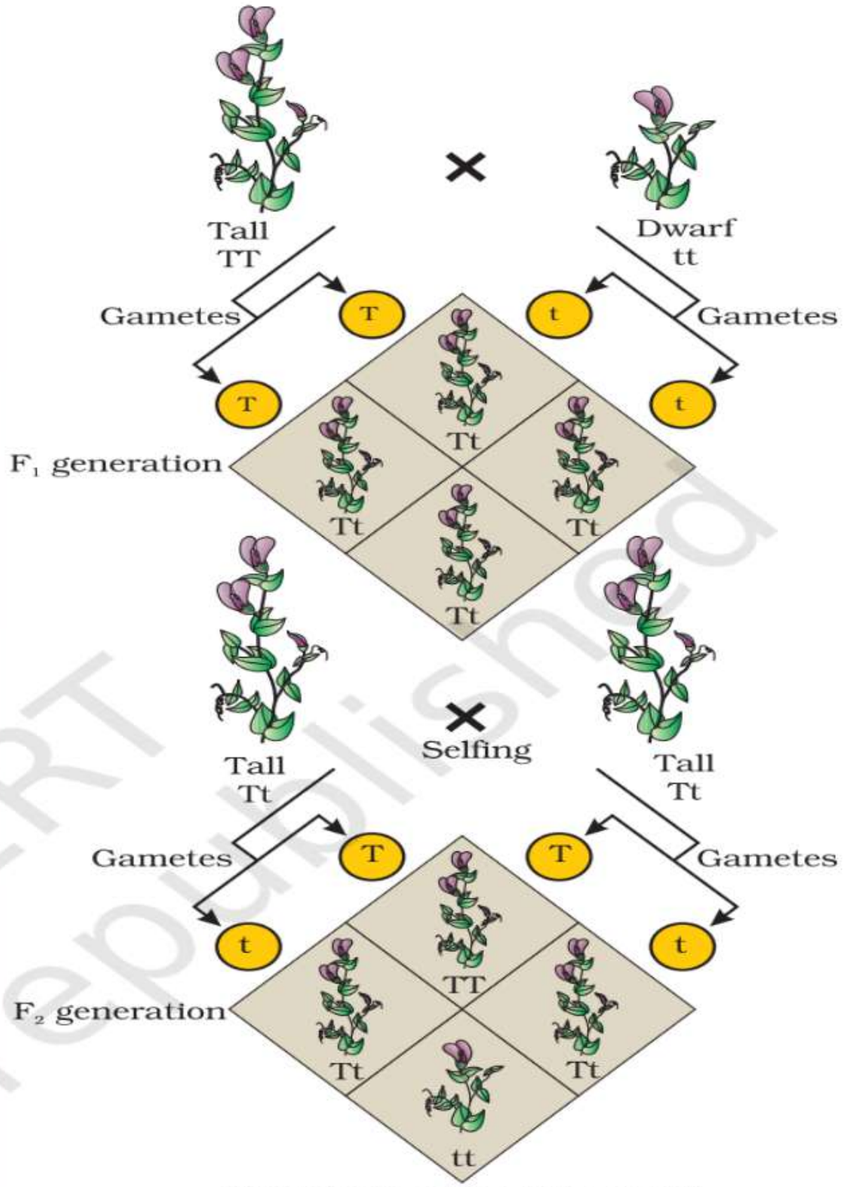
"Punnett Square" "Graphical representation"

zygotic combinations

Phenotypic ratio = 3 : 1

Genotypic ratio = 1 : 2 : 1


Petal
Stigma
Anther
Stamen
Carpel
Removal of anthers
(Emasculation)
♀
Parent
♂
Parent
Transfer of pollen
(Pollination)

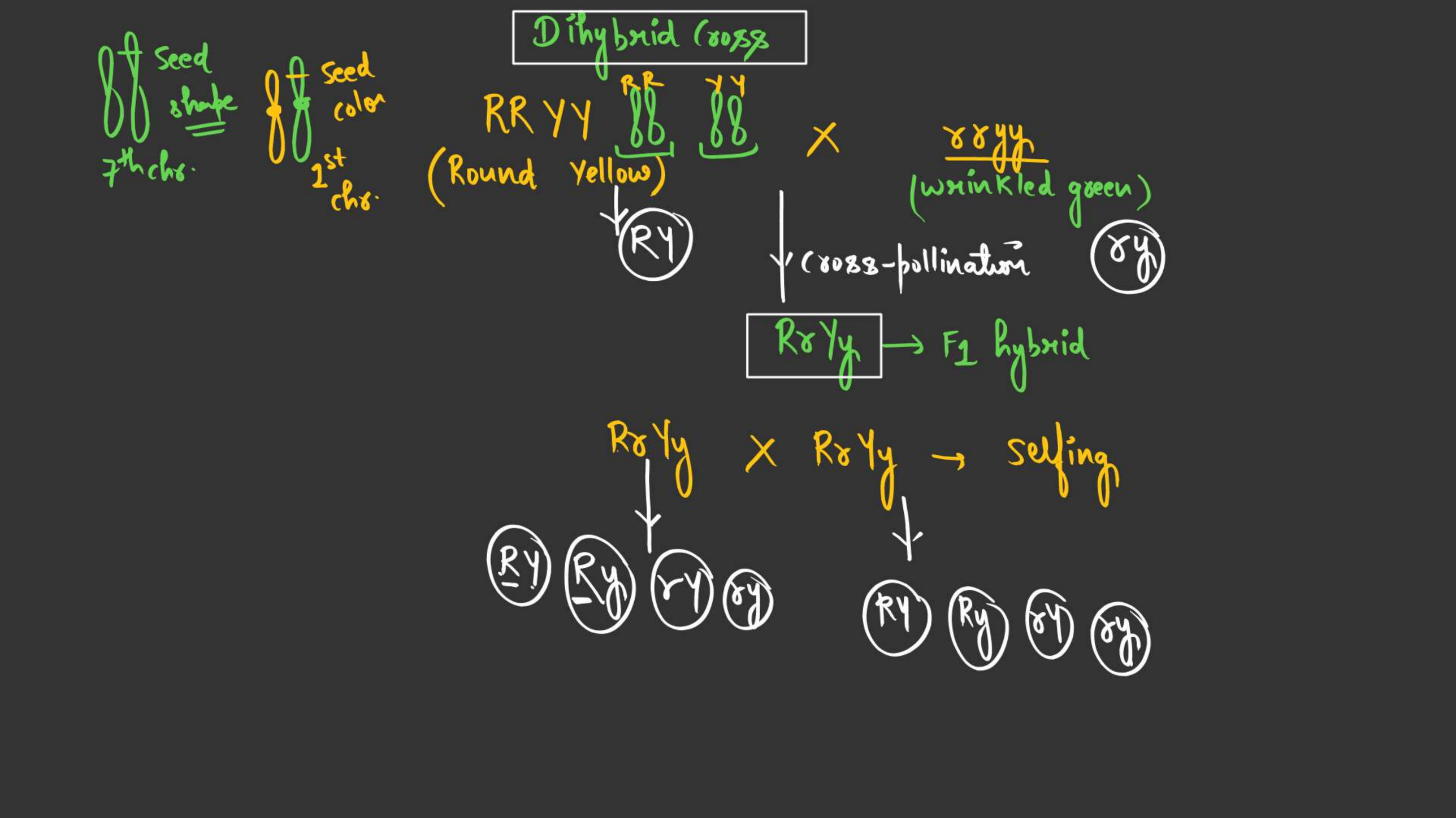


Phenotypic ratio : tall : dwarf
3 : 1
Genotypic ratio : TT : Tt : tt
1 : 2 : 1

F_2 - generation

Phenotypes = ② < Tall, Dwarf (8≡)

Genotypes = ③ TT, Tt, tt


Dihybrid cross
Seed shape
7th chr.
Seed color
1st chr.
RR YY
RR
YY
(Round Yellow)
×
rr yy
(wrinkled green)
RY
ry
cross-pollination
Rr Yy
→ F1 hybrid
Rr Yy × Rr Yy → selfing
RY
Ry
rY
ry
RY
Ry
rY
ry

Seed shape

Fork method

(Genotypes) (Phenotypes)

1 RR

- YY 1 = RR YY ① → Round yellow
- Yy 2 = RR Yy ② → " "
- yy 1 = RR yy ① → Round green

2 Rr

- YY 1 = Rr YY ② → Round yellow
- Yy 2 = Rr Yy ④ → " "
- yy 1 = Rr yy ② → Round green

1 rr

- YY 1 = rr YY ① → wrinkled Yellow
- Yy 2 = rr Yy ② → wrinkled Yellow
- yy 1 = rr yy ① → wrinkled green

F2 generation

Genotypes = 9

Phenotypes = 4

Genotypic ratio

1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

Phenotypic ratio

Round yellow : Round green : wrink yellow : wrink green

9 : 3 : 3 : 1

Laws of inheritance

① **Law of Dominance** → Not Universal

→ Based on Monohybrid Cross.

fails at
* Codominance
* Incomplete dominance.

4.2.1 Law of Dominance

(i) Characters are controlled by discrete units called **factors**.

(ii) Factors occur in pairs.

(iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

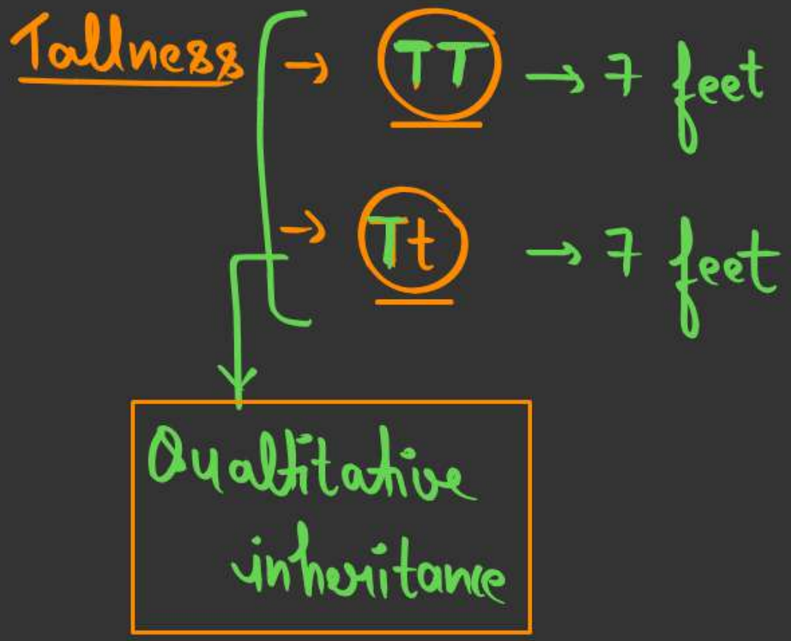
The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F_1 and the expression of both in the F_2. It also explains the proportion of 3:1 obtained at the F_2.

Imp

Dominant trait can express itself both in heterozygous & homozygous condition

Recessive trait can express itself only in homozygous condition

Dwarfness ⇒ (tt)



Q. Which of the following trait can express itself only in homozygous condition?

Recessive trait

a) Yellow seed → (D)

b) Axial flower → (D)

c) Yellow pod → (R)

d) Violet flower → D

Law of Segregation

Based on : Monohybrid cross

Diploid individual

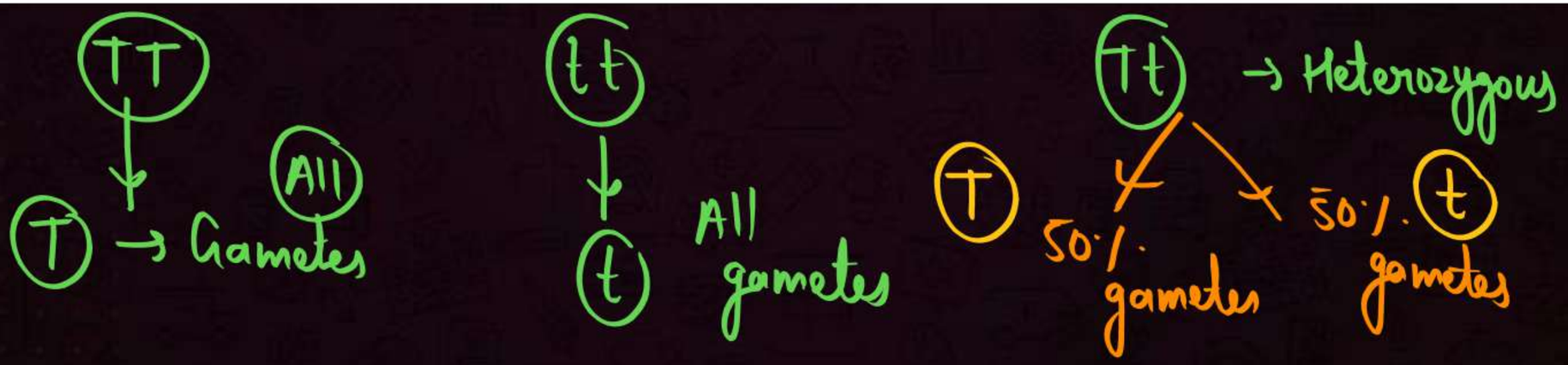
Gamete formation → Meiosis

Segregation of traits

(segregation of chromosomes & genes (factors))

4.2.2 Law of Segregation

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.

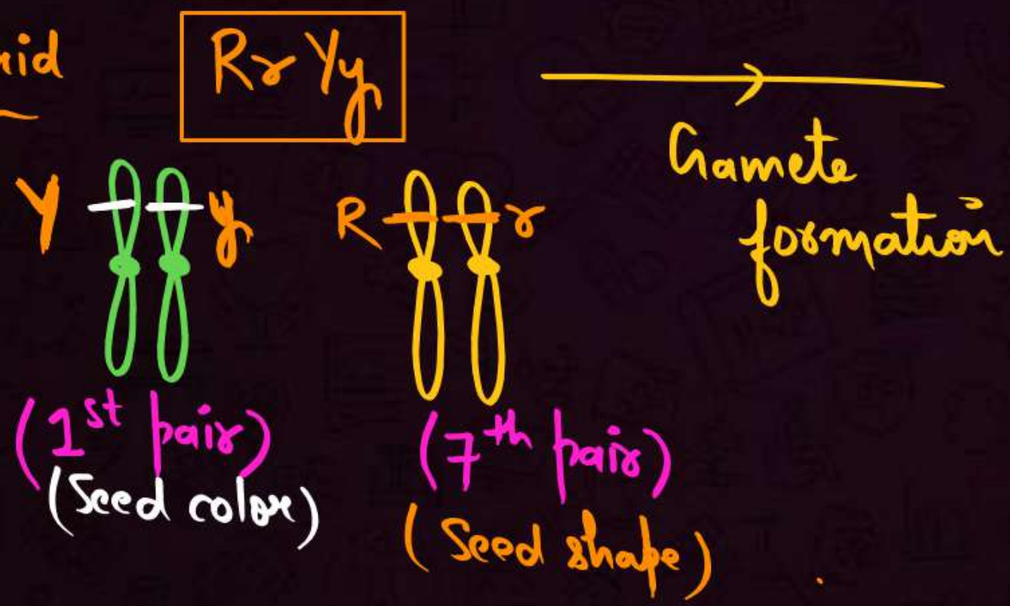


Law of independent Assortment

- Based on Dihybrid Cross
- Not Universal
  - Fails at "Linkage"

In a dihybrid cross

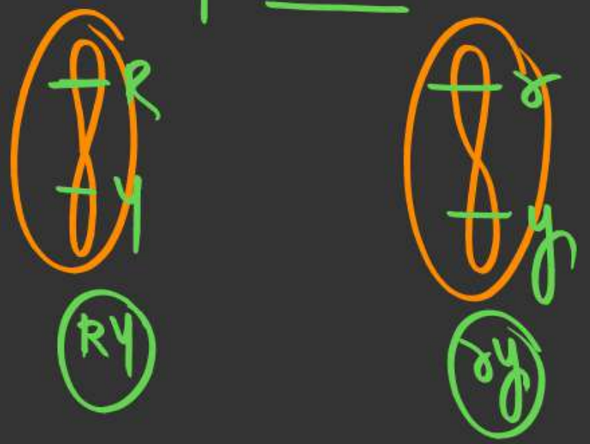
F_1 hybrid — RrYy



Suppose: These (2) genes were present on same chromosomes.

(F1 hybrid) (RrYy)

↓ Gamete



No independent Assortment

linkage

8 * 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

F_1 hybrid × Either of the Parent

Outcross

F_1 × Dominant parent

For ex → (Tt) × TT

F_1

"Definition"

Test Cross

F_1 × Recessive parent

(Tt) × (tt)

F_2 gen. → F_2 gen × Recessive parent

Mendel

F2 gen. → Tall ← TT, Tt

(Used Test cross)

Test-cross → utility Used to check homozygosity / heterozygosity of the individual showing dominant phenotype

Tall (phenotype)

Plant (Unknown genotype) × tt

TT × tt → Tt All tall

Tt × tt → T, t, t → Tt : tt

Result

All offsprings (seed) Tall

(Genotype → TT)

Result

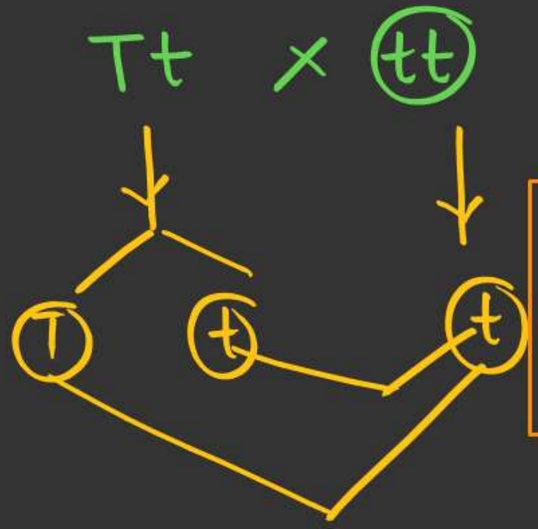
offsprings

50% → Tall

50% → dwarf

Genotype → tt

Monohybrid Test Cross



P·R = G·R = 1:1

Genotypes = Phenotypes

Tt → Tall

tt → dwarf

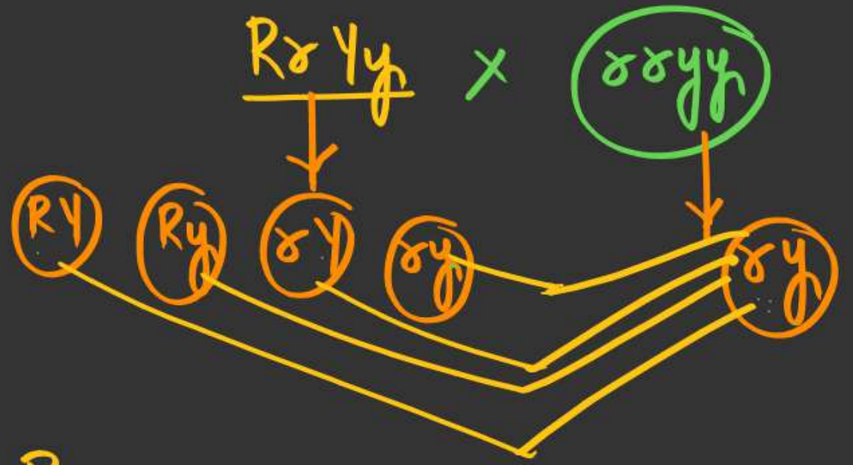
Phenotypes → 2

Genotypes → 2

P·R = Tall : dwarf
1 : 1

G·R = Tt : tt
1 : 1

Dihybrid Test Cross



RrYy Rryy rrYy rryy → Genotypes

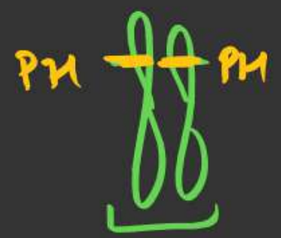
Round yellow | Round green | wrinkled yellow | wrinkled green

Phenotypes = Genotypes = 4

Phenotypes

P·R = G·R = 1:1:1:1

Types of Gametes



2^n n = No. of heterozygous conditions

Aa BB cc

Genotype

ABc

aBc

$2^n = 2^1 = 2$

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

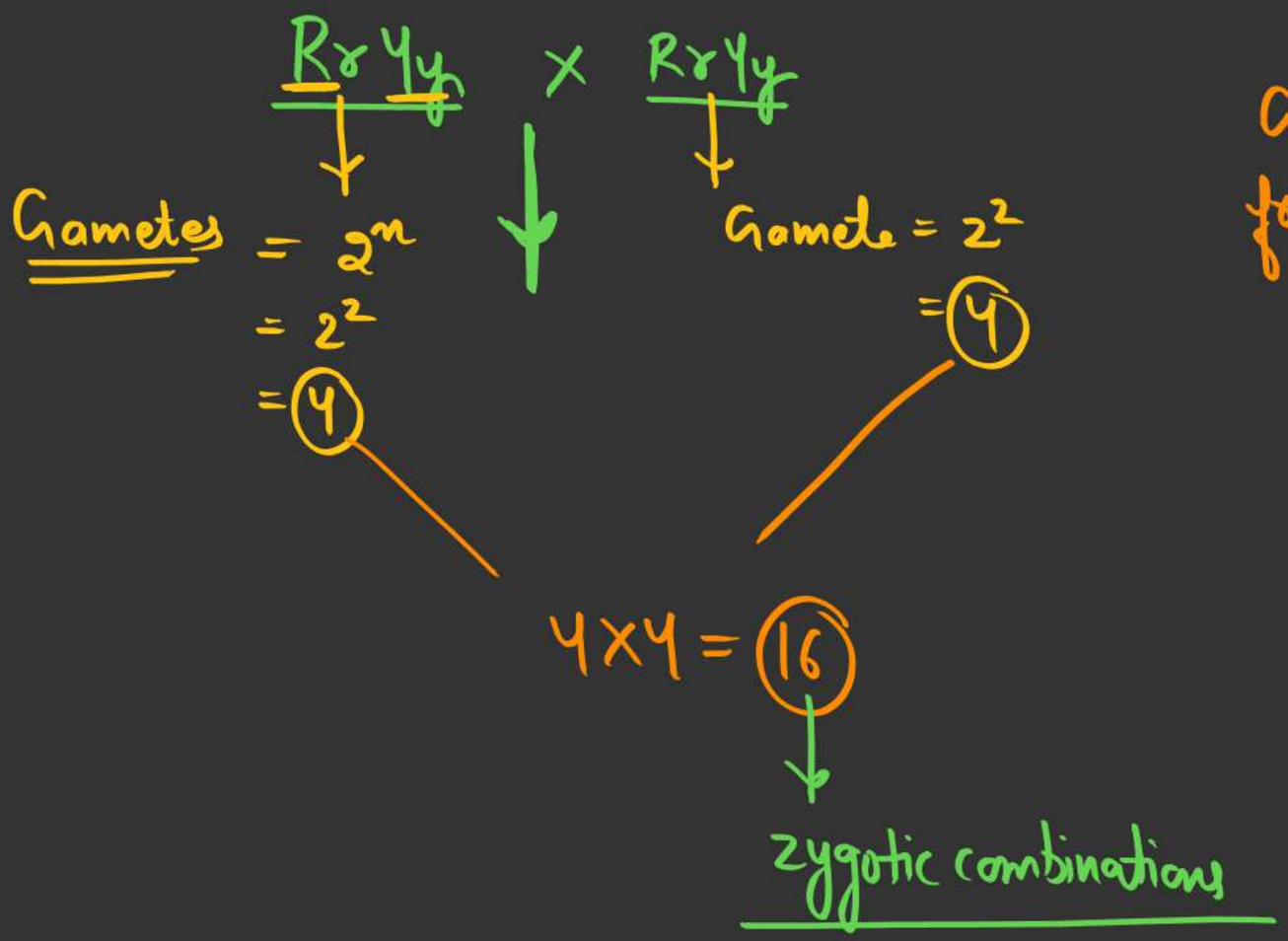
Ans. Aa BbCc

$2^n = 2^3 = 8$

Ex F_1 = $R_r Y_y$

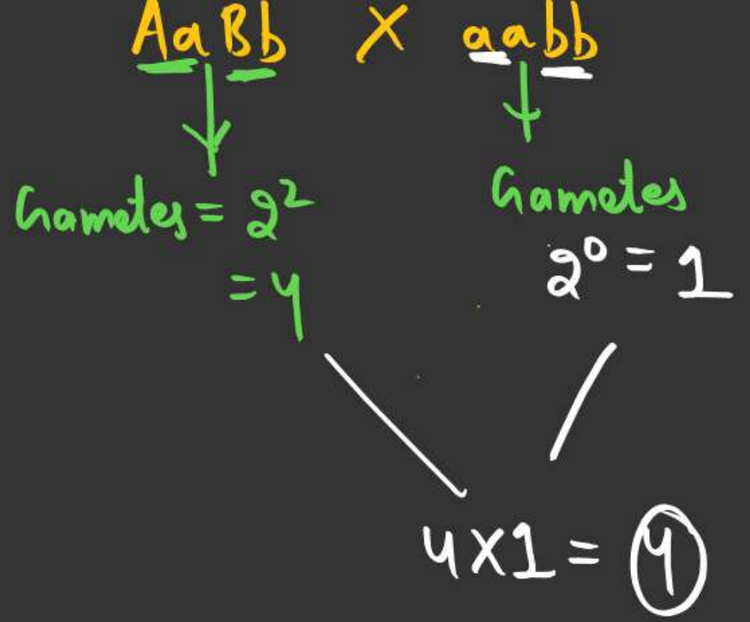
$2^2 = 4$

Number of offsprings / zygotic combinations



Formula:

Gametes from Parent$_1$ × Gametes from Parent$_2$



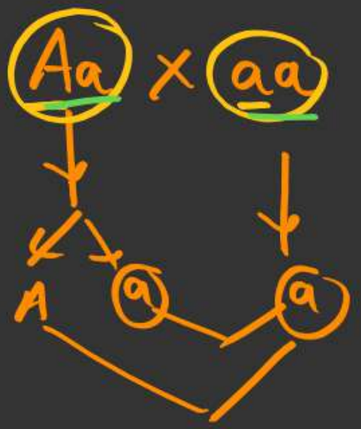
Frequency method

AA × aa

↓

Aa → All

→ 100% (1)



Aa → 1/2

aa → 1/2

Aa × AA

Aa → 1/2

AA → 1/2

Aa × Aa

AA → 1/4

Aa → 2/4

aa → 1/4

	A	a
A	AA	Aa
a	Aa	aa

AA × AA

↓

AA → 1

aa × aa

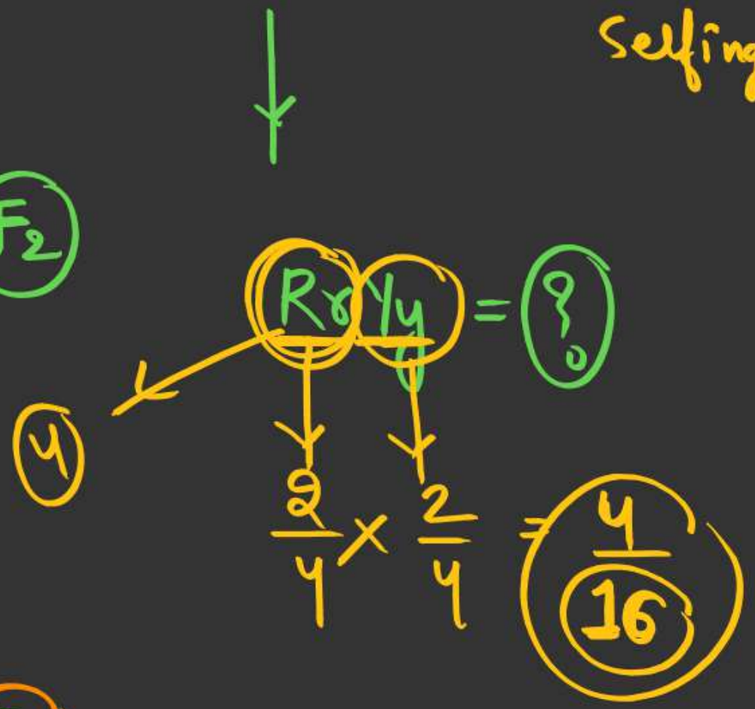
↓

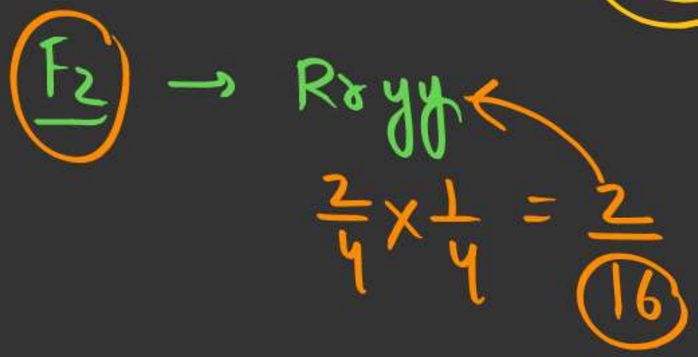
aa → 1

Dihybrid cross :

RrYy × RrYy → F_1

Selfing





Rr × Rr

RR → 1/4

Rr → 2/4

rr → 1/4

Yy × Yy

YY → 1/4

Yy → 2/4

yy → 1/4

F_2 = rryy

1/4 × 1/4 = 1/16

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

Calculate the proportion of offsprings given below

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

(b) dwarf and green

$Tt \times Tt$

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

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

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

$Yy \times yy$

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

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

(a) Tall and green

$TTyy + Ttyy$

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

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

$TtYy \times Ttyy$

Gametes $= 2^2 = 4$

Gametes $= 2^1 = 2$

(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) $RrYy \times RrYy$

$Rr \times Rr$ < $RR \to \frac{1}{4}$, $Rr \to \frac{2}{4}$, $rr \to \frac{1}{4}$

$Yy \times Yy$ < $YY \to \frac{1}{4}$, $Yy \to \frac{2}{4}$, $yy \to \frac{1}{4}$

F_2 gen.

800

offsprings / seed → Round green (yy) = $\frac{1}{4}$

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

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

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

16 → 3

1 → $\frac{3}{16}$

Total seed = $\frac{3}{16} \times 800^{50}$

= 150 seeds

Exceptions of Mendelism

a) Incomplete dominance

b) Complete dominance

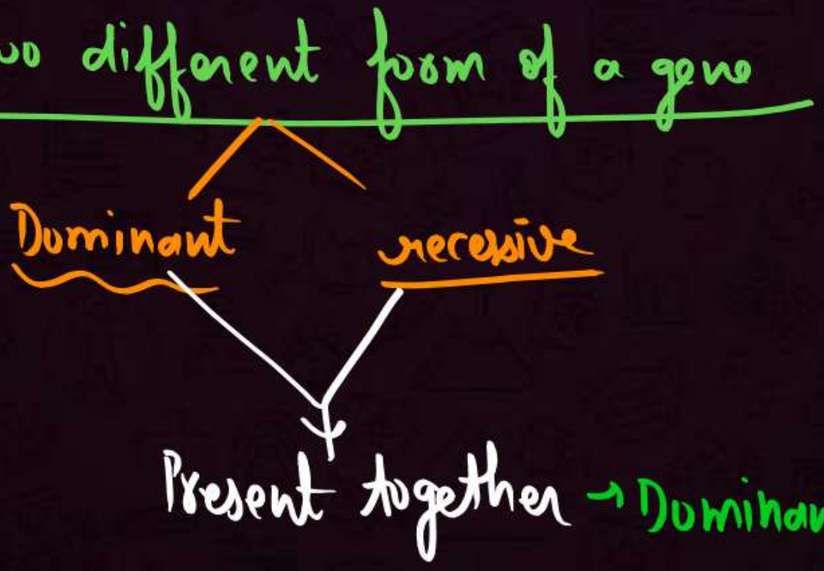
c) Multiple alleliism.

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



Dogflower

Red (RR) × rr (white)

↓

F1 — Rr (Pink)

Rr × Rr → Selfing

	R	r
R	(Red) RR	(Pink) Rr
r	(Pink) Rr	(white) rr

F2

In F_2

Genotypes = 3

Phenotypes = 3

P·R = Red : Pink : white

1 : 2 : 1

G·R = RR : Rr : rr

1 : 2 : 1

G·R = P·R

Co-dominance

↓

Two different forms of a same gene
- A → Dominant
- B → Dominant

→ Present together → Express equally

Blood group gene
- I^A → A-bld grp
- I^B → B bld grp

$I^A I^B$ → AB Blood group

↓

Example of Co-dominance

Co-dominant alleles

* 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 → Blood group system in humans → ABO blood system

Blood group - gene

- I^A (Dominant form) → Formation of A-antigen (RBC membrane)
- I^B Dominant form → B-antigen
- I^o/i (Recessive form) → No antigen

* Can't be detected in an individual.

* It is a trait of Population

Genotype

$I^A I^A$

$I^A I^O$

Phenotype (Blood group)

(A)

$I^B I^B$

$I^B I^O$

(B)

$I^A I^B \rightarrow AB$

$I^O I^O$ | ii $\rightarrow O$

Population

Phenotypes → (4)

Genotypes → (6)

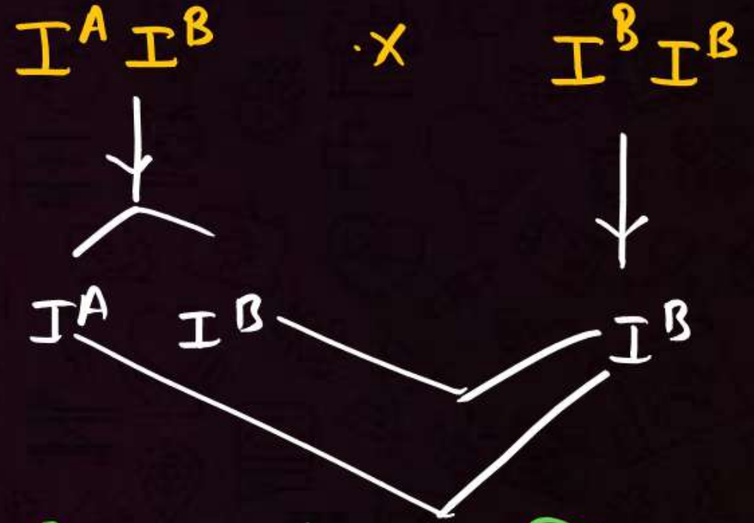
Q What will be the possible blood group in children from the parents B and AB blood groups?

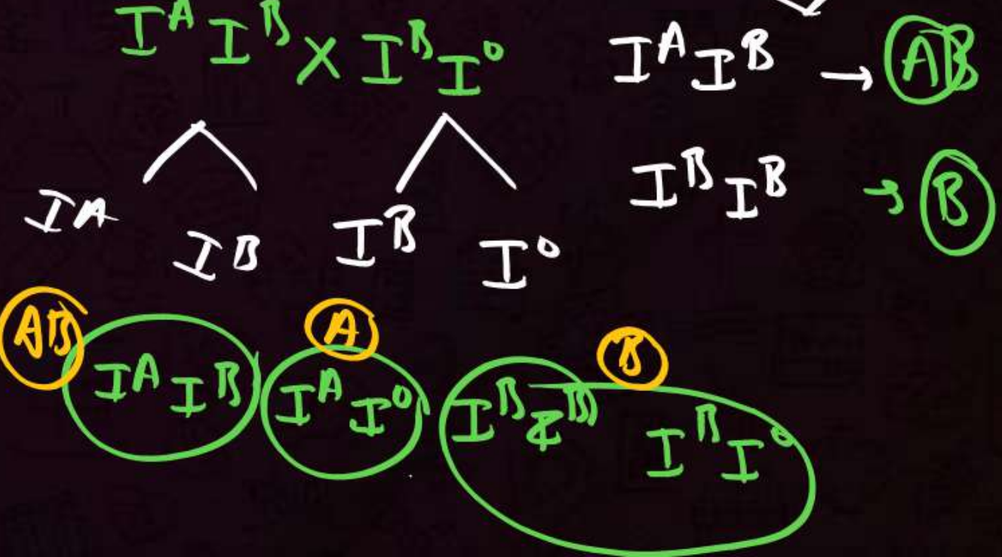
a) A, O

b) A, B, AB and O

c) A, B, AB

d) B, O





PLEIOTROPY

A pleiotropic genes affects / controls several phenotypes or traits

Example: a) Hb^S gene (Mutated gene) (Causes Sickle anaemia)

b) Mutated gene causing disease PhenylKetonuria

c) B- gene → Starch Branching enzyme gene

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

B-gene → Pisum sativum

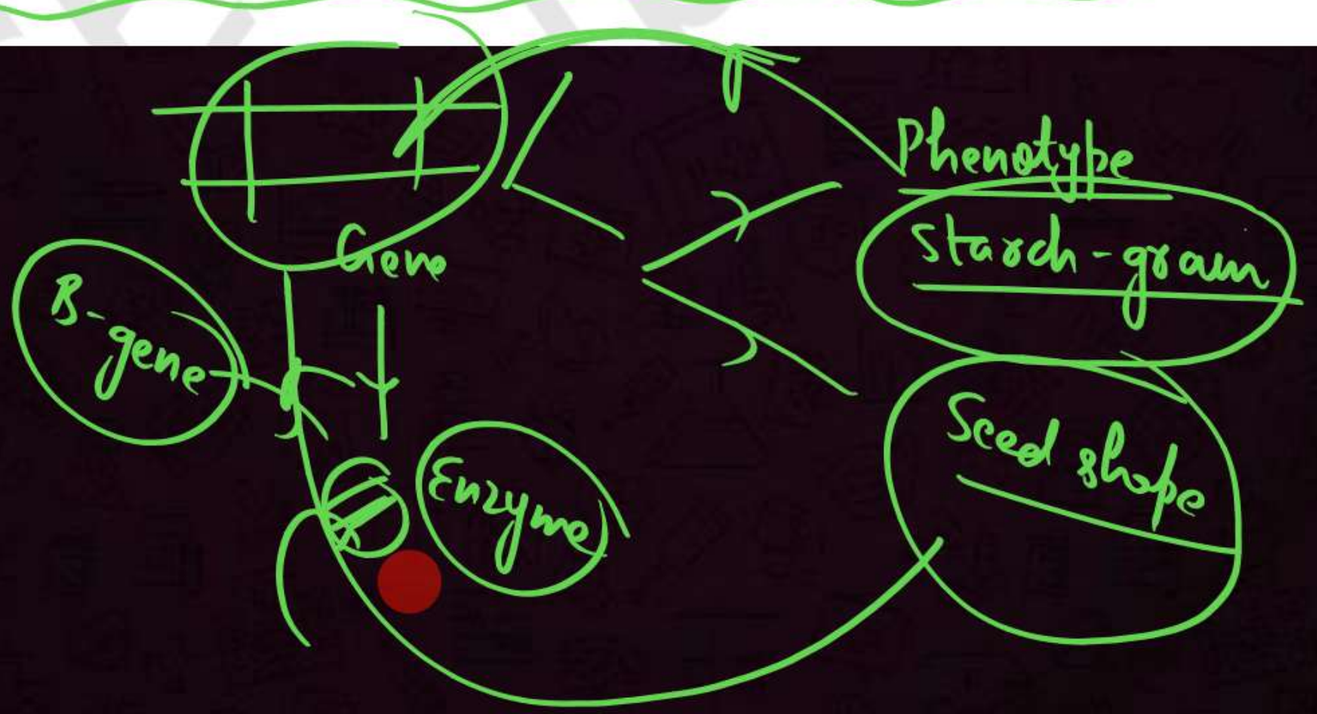
Controls (2) Phenotypes → Starch grain synthesis, Seed-shape

B-gene	Starch-grain Size	Seed shape
BB	Long	Round
Bb	Intermediate (Incomplete dominance)	Round (Complete dominance)
bb	Small	wrinkled

* when B-gene controls starch-grain 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

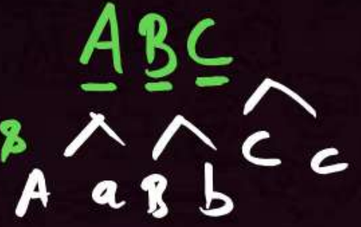
* Quantitative inheritance
  the expression is affected by number of dominant alleles

* Effect of each dominant allele is additive

Ex → Skin color in Humans

→ Height in Humans

→ Intelligence in Humans

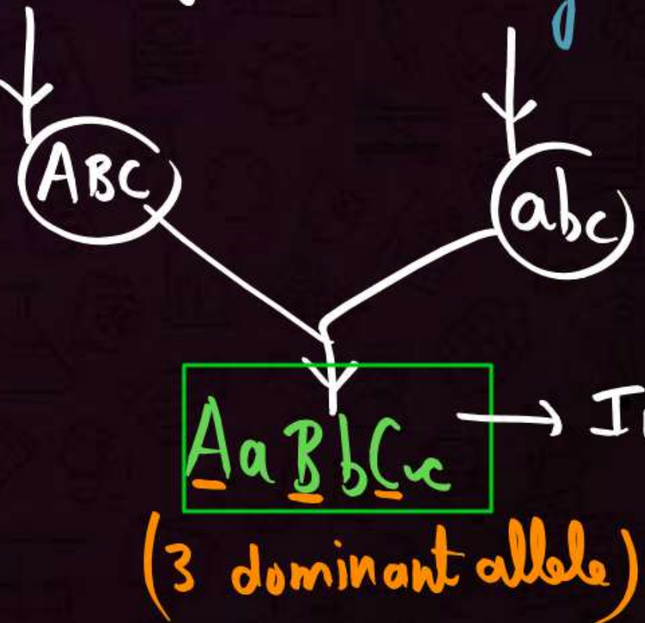


Skin-color → Controlled ③ genes (Polygenes)

Total alleles → ⑥

AABBCC ⑥ dominant alleles (Very dark) (Negro)

× aabbcc (0 dominant allele) very light (Albino)



→ Intermediate (Mulatto)

Rediscovery of Mendel's work

In 1900

- Hugo de Vries
- Carl Correns
- Tschermark

* 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)

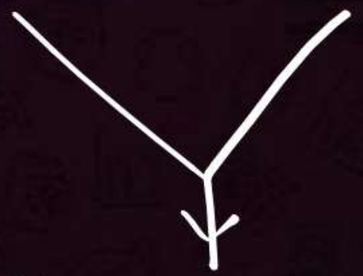
d) His concept of factors → "discrete units"
↓
"do not blend"
was not accept by his contemporaries (Darwin)

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

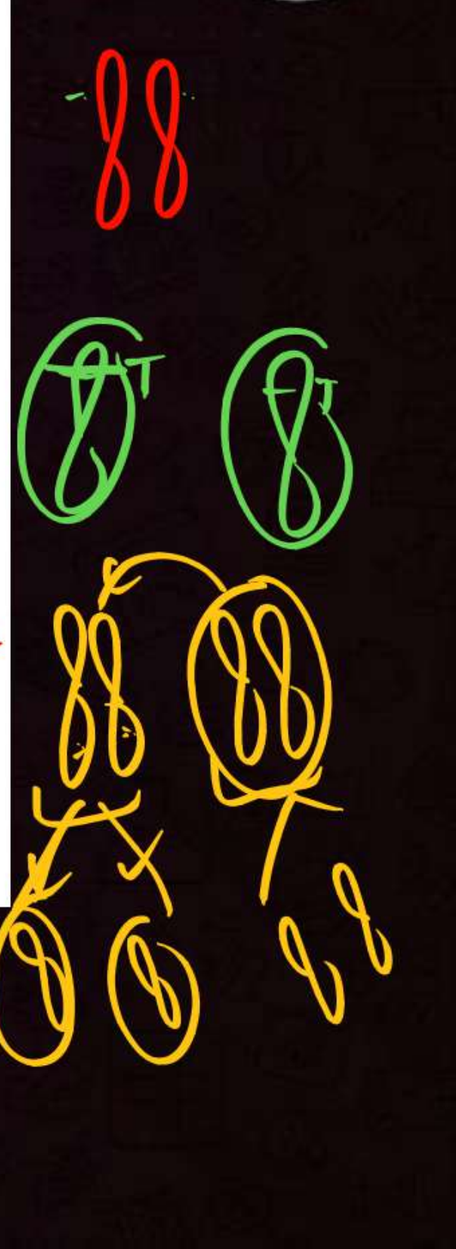


United the Knowledge of Mendel's work and Electron microscopy studies

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

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

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



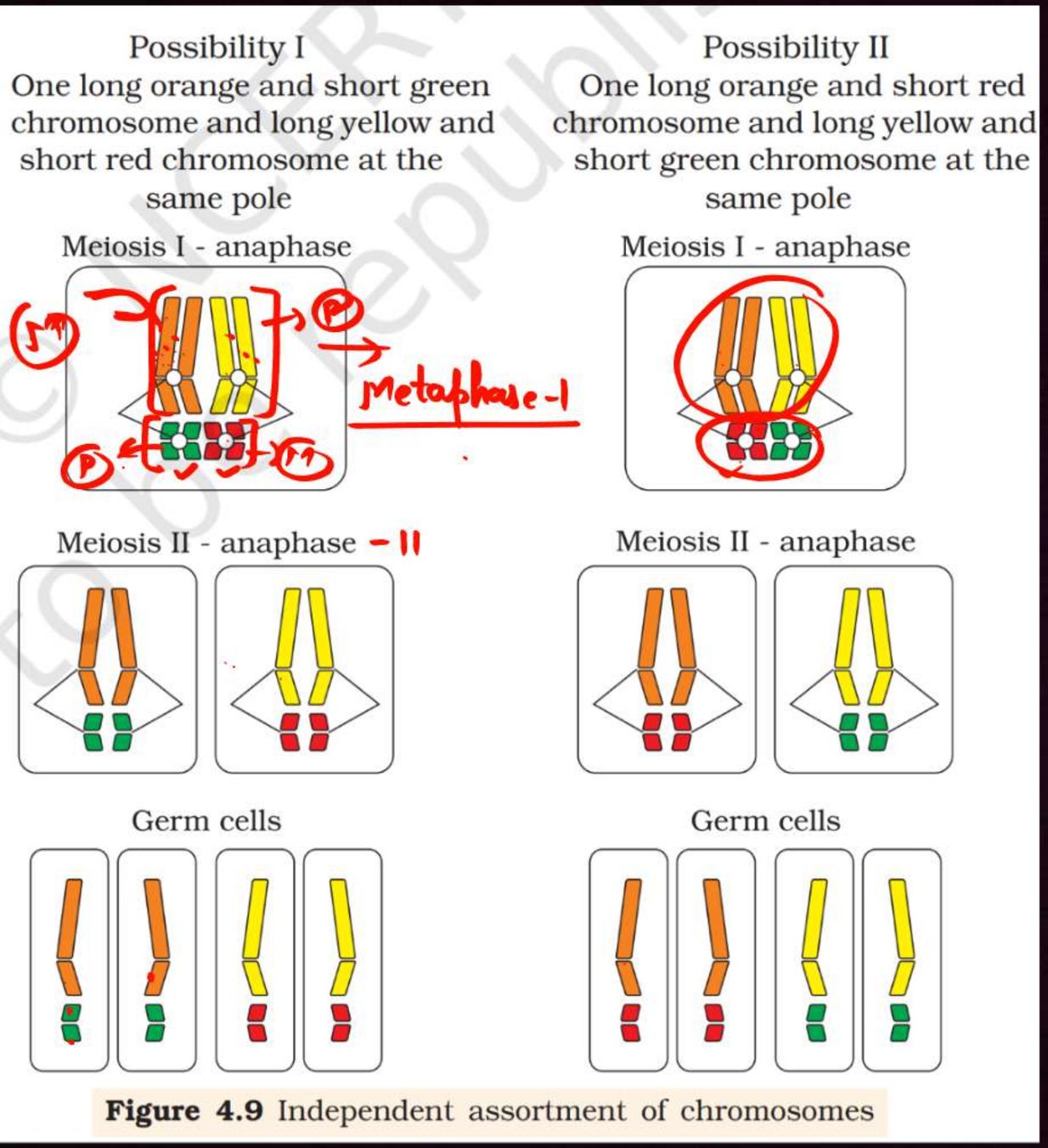


Figure 4.9 Independent assortment of chromosomes

Experimental Genetics → Father

Thomas Hunt Morgan

Explaind Linkage ★ ★

Model Organism

Drosophila melanogaster

(Fruitfly)

Gave

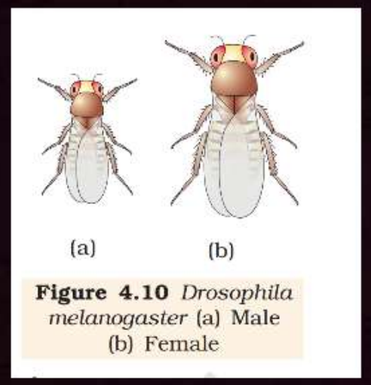
Experimental Verification of Chromosomal theory of inheritance.

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

Why Morgan chose Drosophila

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

Male → shorter
Female → longer.



(a) (b)

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

5. Males have Heteromorphic sex-chromosomes.

Females — XX

Males — XY

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

Drosophila

Diploid organism

$2n = 8$ → (4) pairs of chromosomes

- Males: 6 + XY
  - 6 (3 pair) (Autosomes)
  - XY: Allosomes (sex chromosomes)
- 6 + XX

LINKAGE

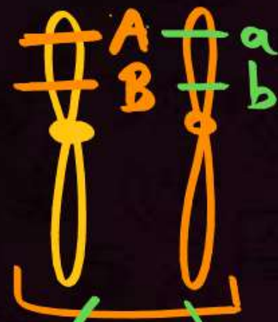
→ All the genes present on same chromosome are physically associated & are inherited together

This phenomenon is called as linkage.

Linkage

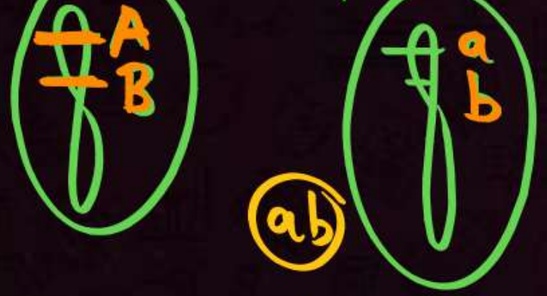
Complete Linkage

AaBb



"No crossing over"

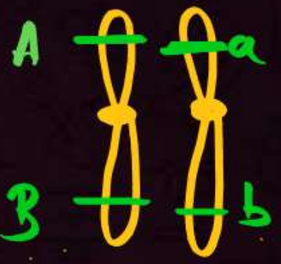
Gametes



(AB) (ab)

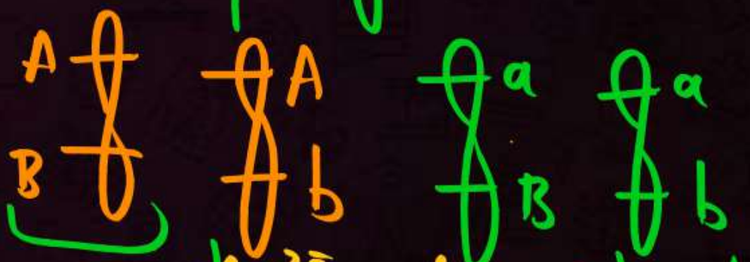
* No recombinants
* 100% Parental type

Incomplete Linkage



→ Crossing-over can occur and can form recombinants

④ gametes



more (25%) Parental type

Less 25% Less 25% Recombinants

(25%) Parental type

(D) allele → (3) ways: T, t^+, +

(R) allele → t

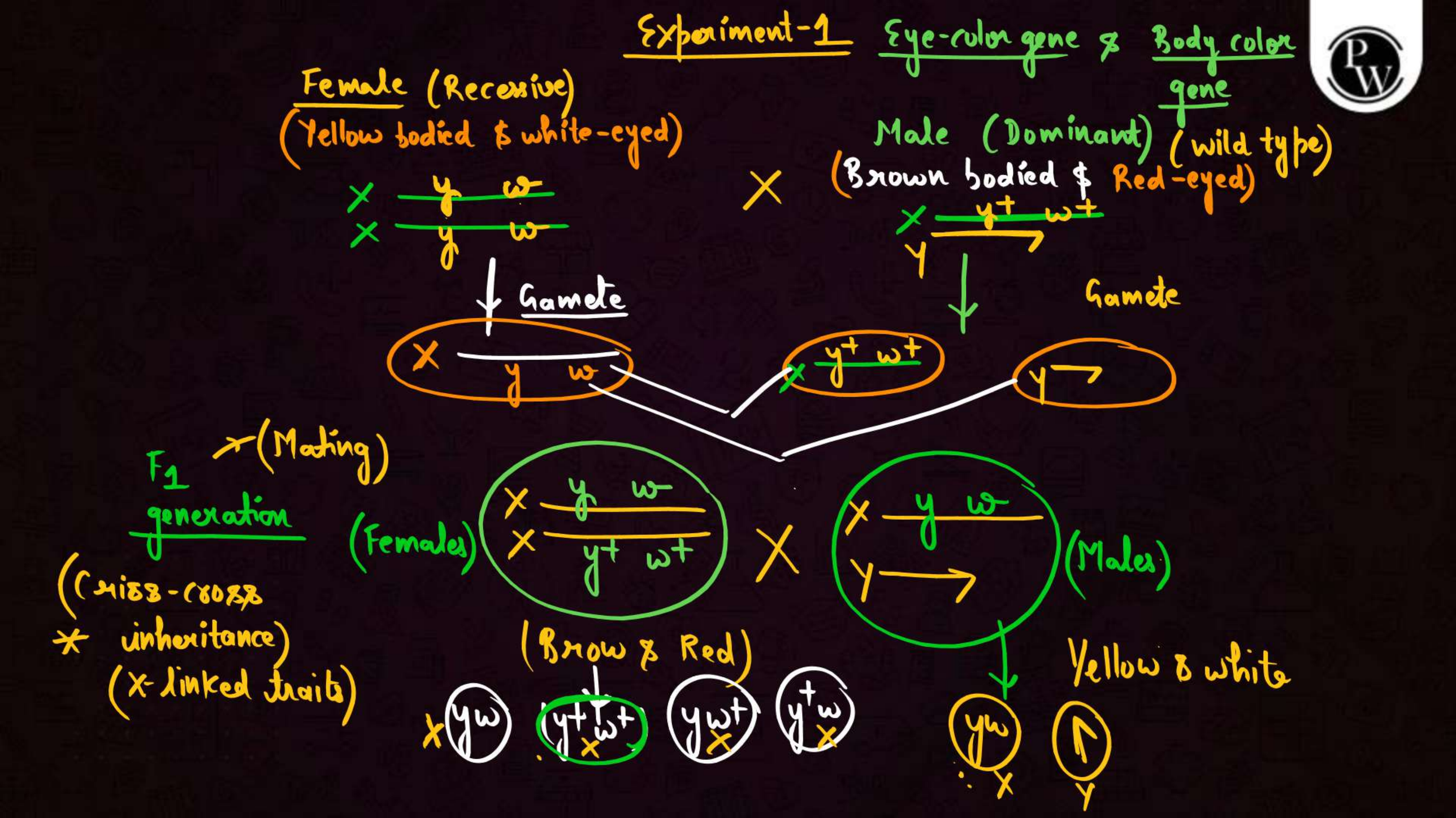
Morgan Experiment → "Sex-linked inheritance"

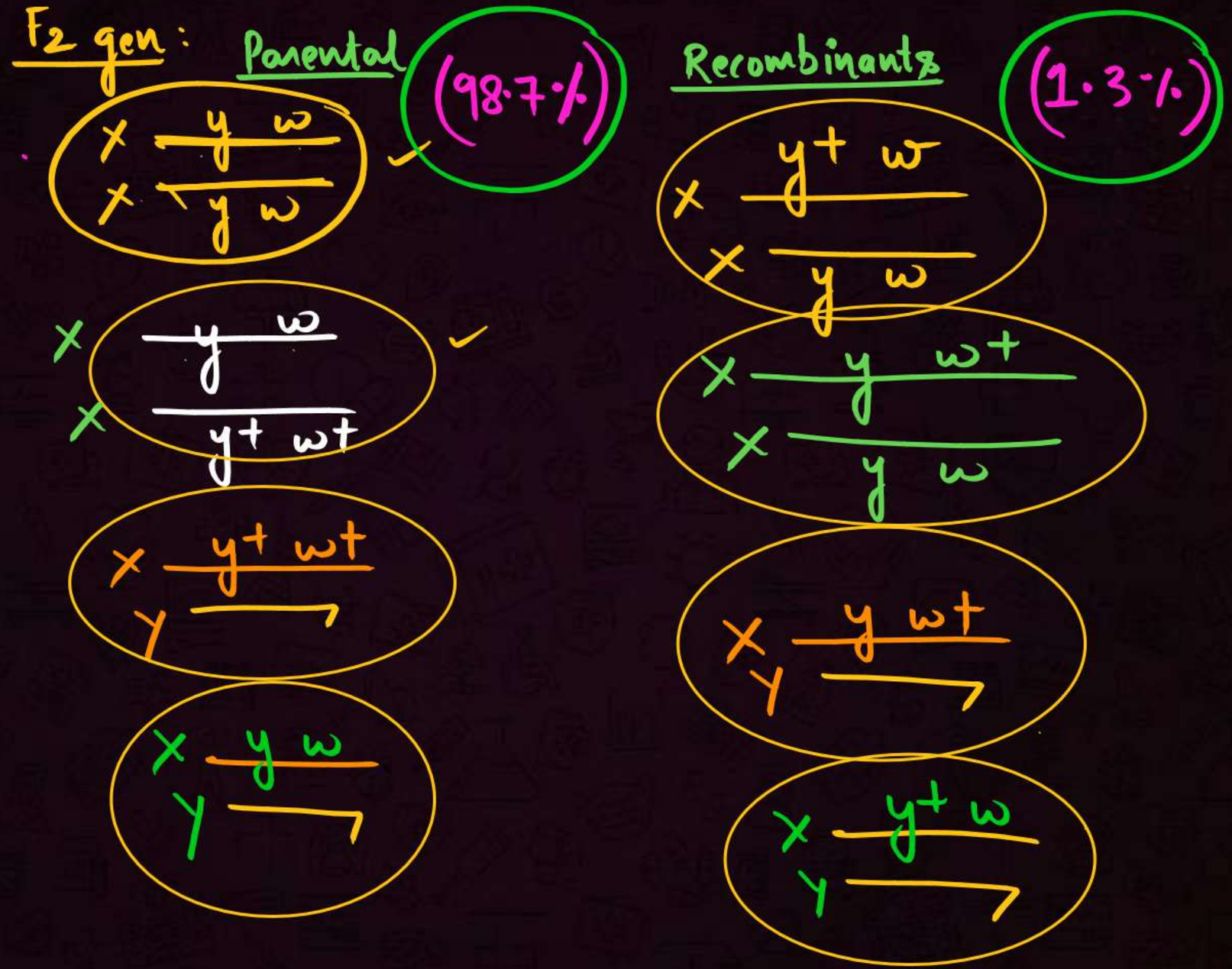
Inheritance of genes present on sex-chromosome

Drosophila

"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)


Experiment-1
Eye-color gene & Body color gene
Female (Recessive)
(Yellow bodied & white-eyed)
Male (Dominant)
(Brown bodied & Red-eyed)
(wild type)
Gamete
Gamete
F1 generation
(Mating)
(Females)
(Males)
(Criss-Cross inheritance)
(X-linked traits)
(Brown & Red)
Yellow & white


F2 gen :
Parental
(98.7%)
Recombinants
(1.3%)
X y w / y w
X y w / y+ w+
X y+ w+ / y w
X y+ w+ / Y
X y w / Y
X y+ w / y w
X y w+ / y w
X y w+ / Y
X y+ w / Y

Experiment-2

Female

$\frac{w \quad m}{w \quad m}$ × $\frac{w^+ \quad m^+}{}$ →

Eye-color gene & wing type gene

"Same-cross"

F_2 gen

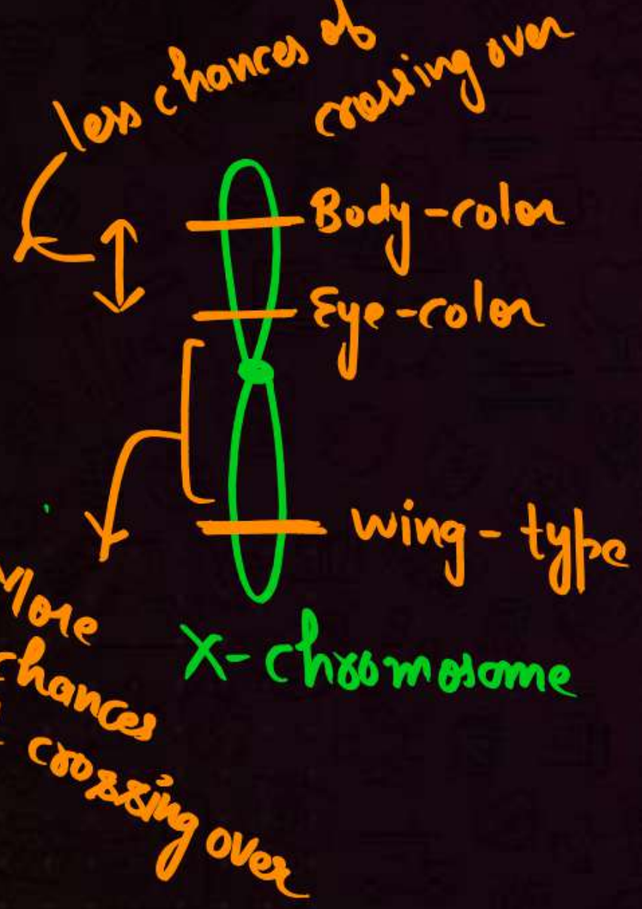
Result

Parental type (62.8%)

Recombinants (37.2%)

Conclusion

Distance between genes on same chromosome $\propto$ Crossing over | Recombination



$\propto \frac{1}{\text{Linkage}}$

Linkage $\propto \frac{1}{\text{crossing over}}$

Chromosomal mapping / Gene-mapping

↓ Alfred Sturtevant (Drosophila)

* Genes are arranged linearly on chromosome.
* The arrangement of genes / Position of genes can be predicted by **Recombination 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) (Pro)

Morgan's Experiment

Eye-color & Body-color

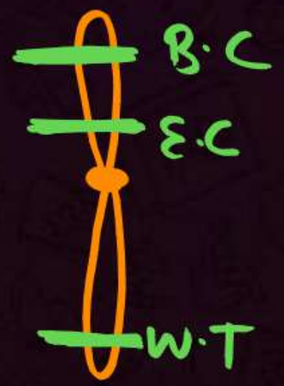
R.F / C.O.V = (1.3%)

* 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%)

Distance b/w genes = (37.2 cM)



Q. Distance between A-B = 3 M.U B-C = 7 M.U

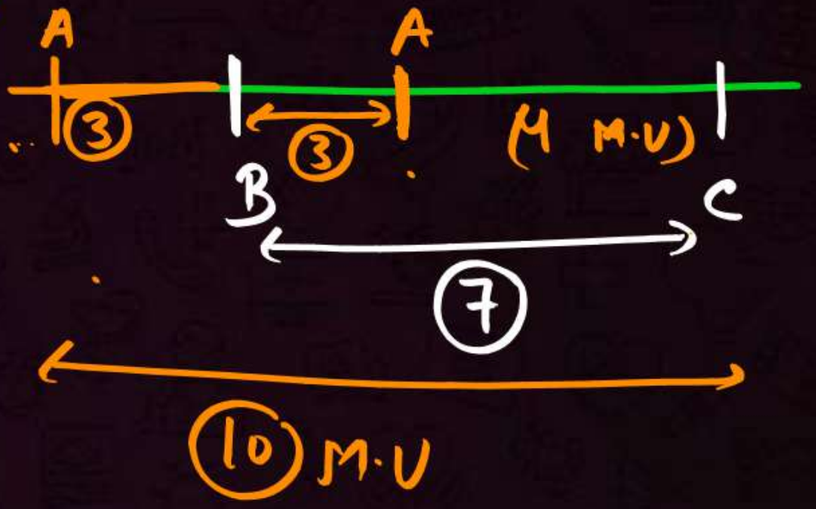
A-C = ?

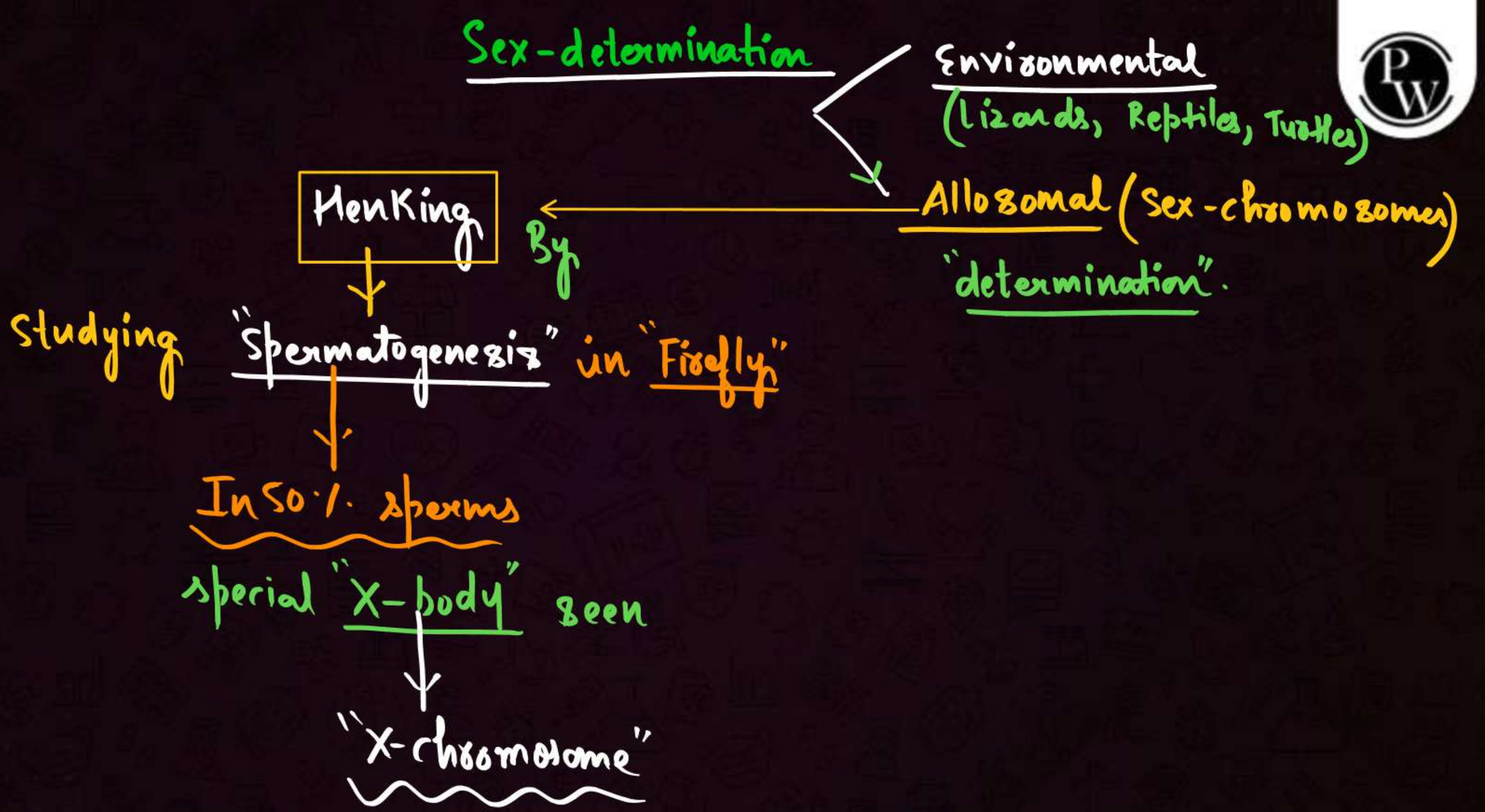
a) 4 M.U

b) 10 M.U

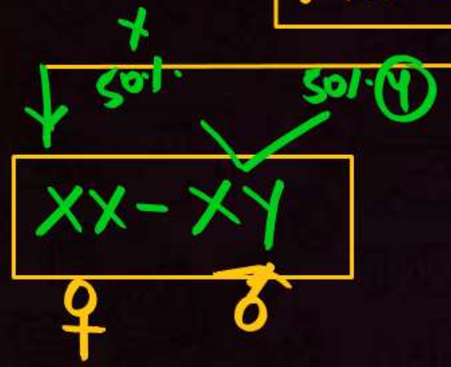
c) 6 M.U

d) Either (a) or (b)




Sex-determination
Environmental
(Lizards, Reptiles, Turtles)
Allosomal (Sex-chromosomes)
"determination".
By
HenKing
Studying
"Spermatogenesis"
in "Firefly"
In 50% sperms
special "X-body" seen
"X-chromosome"

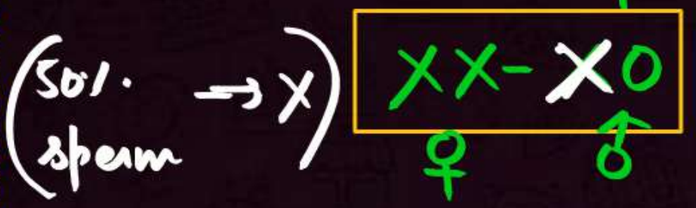
Male Heterogamety → Males decide gender



→ Humans

→ Drosophila

(Sol. → X)



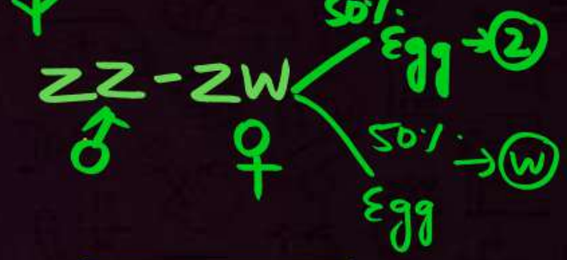
50% sperm (No sex chromosome)

→ Insects

→ Grasshopper

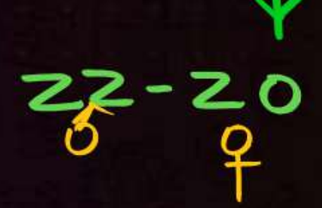
→ Cockroach

Female heterogamety → Females decides gender of progeny



Birds

(Hen)



Butterfly (Moth)

50% Egg — No sex chromosome

50% Egg (Z)

Honey-bee → Haplo-diploidy / Parthenogenesis

Males / Drones → Haploid

16 chromosomes

"Gamete" | "Mitosis"

Male gametes (n)

16 chromosomes

Females (Queen)

Diploid (2n)

32

Gamete | "Meiosis"

Female gamete (n)

16 chromosomes

without fertilisation

Males (Drone) (n)

(Fertilisat^{n}) Male gamete + female gamete

(2n) (Female)

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
- Genomatic mutation
  - change in number of chromosomes
- Chromosomal mutation / Chromosomal abberration
  - change in structure of chromosome

Gene-mutation

Point mutation

change occurs in single base pair.

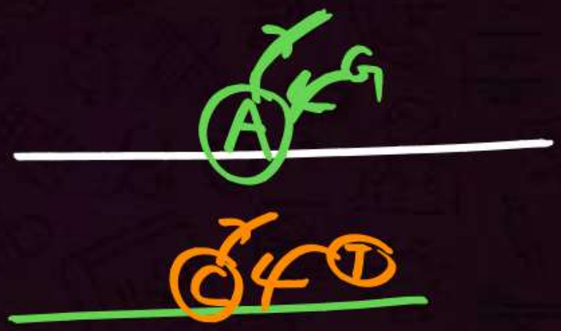
Ex → Sickle Cell Anaemia

Gross mutation

change occurs in many base pairs.

Substitution

Transition



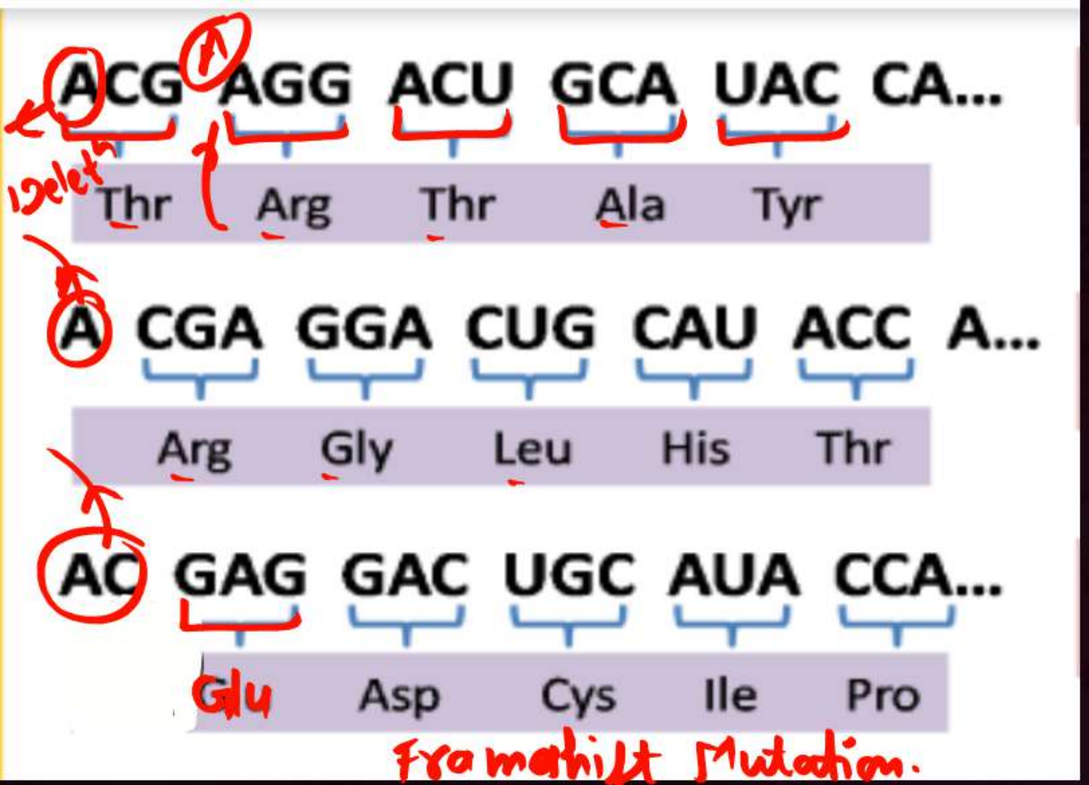
Purine replaced by Purine

Pyrimidine " " Pyrimidine

Transversion

Purine is replaced by pyrimidine (Vice-versa)

(Sickle-cell Anaemia)

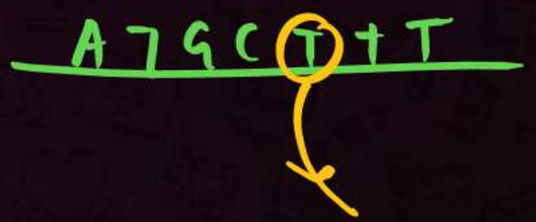


Addition/Insertion

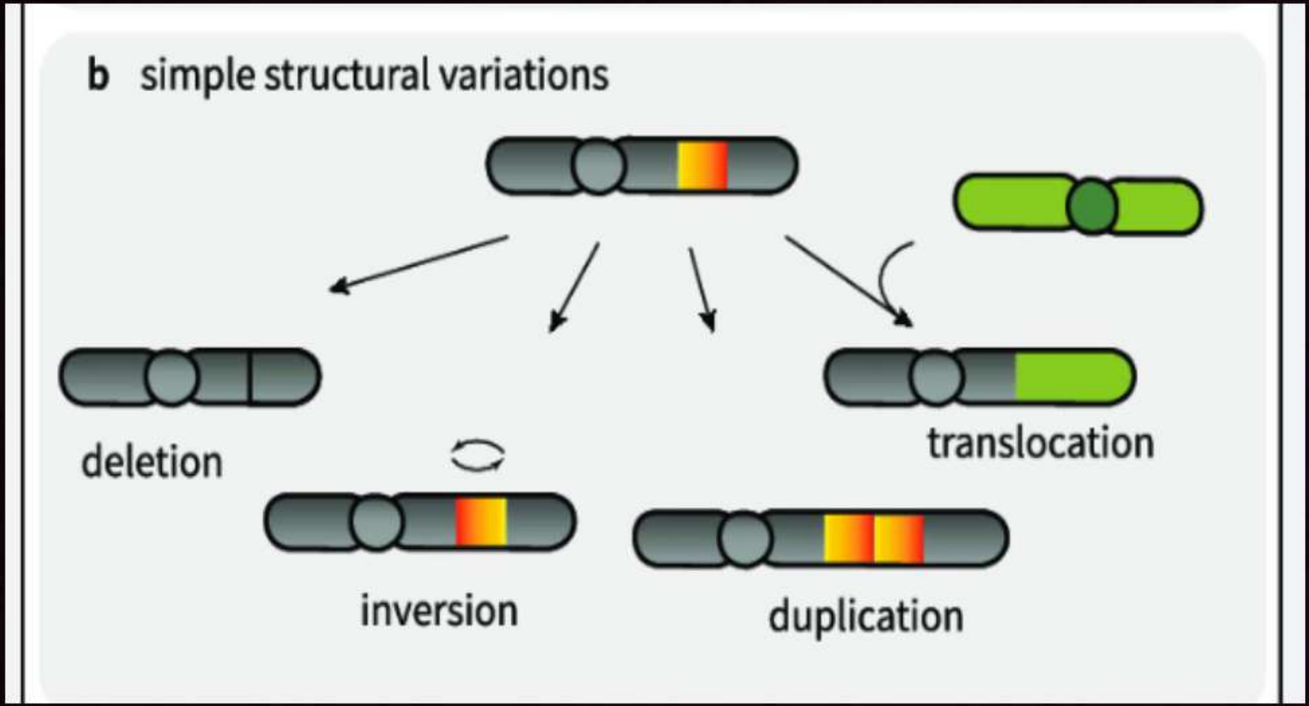
Base/Bases are added

Deletion

Base/Bases → delete



Addition/Deletion → Frameshift Mutation → Thalassaemia



Chromosomal mutations

Genomatic mutation

Aneuploidy

[Change do not occur in complete set of chromosomes but only in a pair or two]

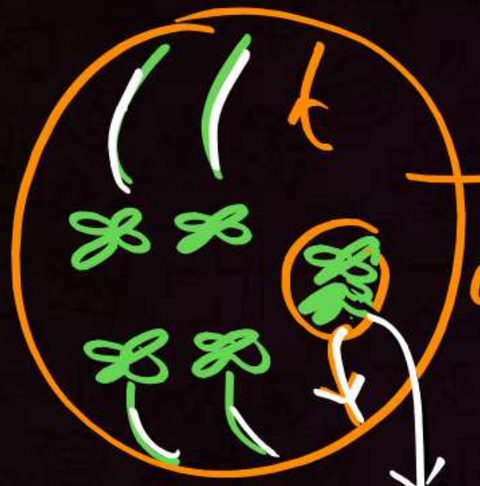
Reason: "Non-disjunction" (do not segregate / separate) of chromosomes during Anaphase

Polyploidy

* when complete set of chromosomes change

$2n \longrightarrow 4n$

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



→ (22) chromosomes
(n-1)

Gamete

→ (24) chromosomes
(n+1)

3rd pair

Gametes (n = 23)

Haploid

Abnormal gamete (n-1) (22) × Normal gamete (n) (23)

(2n-1) → (45) chromosomes

Abnormal gamete (n+1) × Normal gamete (n)

(2n+1) → (47) chromosomes

Aneuploidy

Hyperploidy (Increase)

Trisomy

$(2n+1)$ → 47 chromosomes

→ Down Syndrome

→ Klinefelter syndrome

Hypoploidy (Decrease)

$(2n-1)$ → 45 chr.

MONOSOMY

Ex → Turner Syndrome

Genetic disorders

Non - Mendelian disorders

Cause
- Chromosomal mutation
- Genomatic mutation

Ex
- Down Syndrome
- Klinefelter syndrome
- Turner Syndrome

Mendelian disorders

Cause → "Gene mutation"

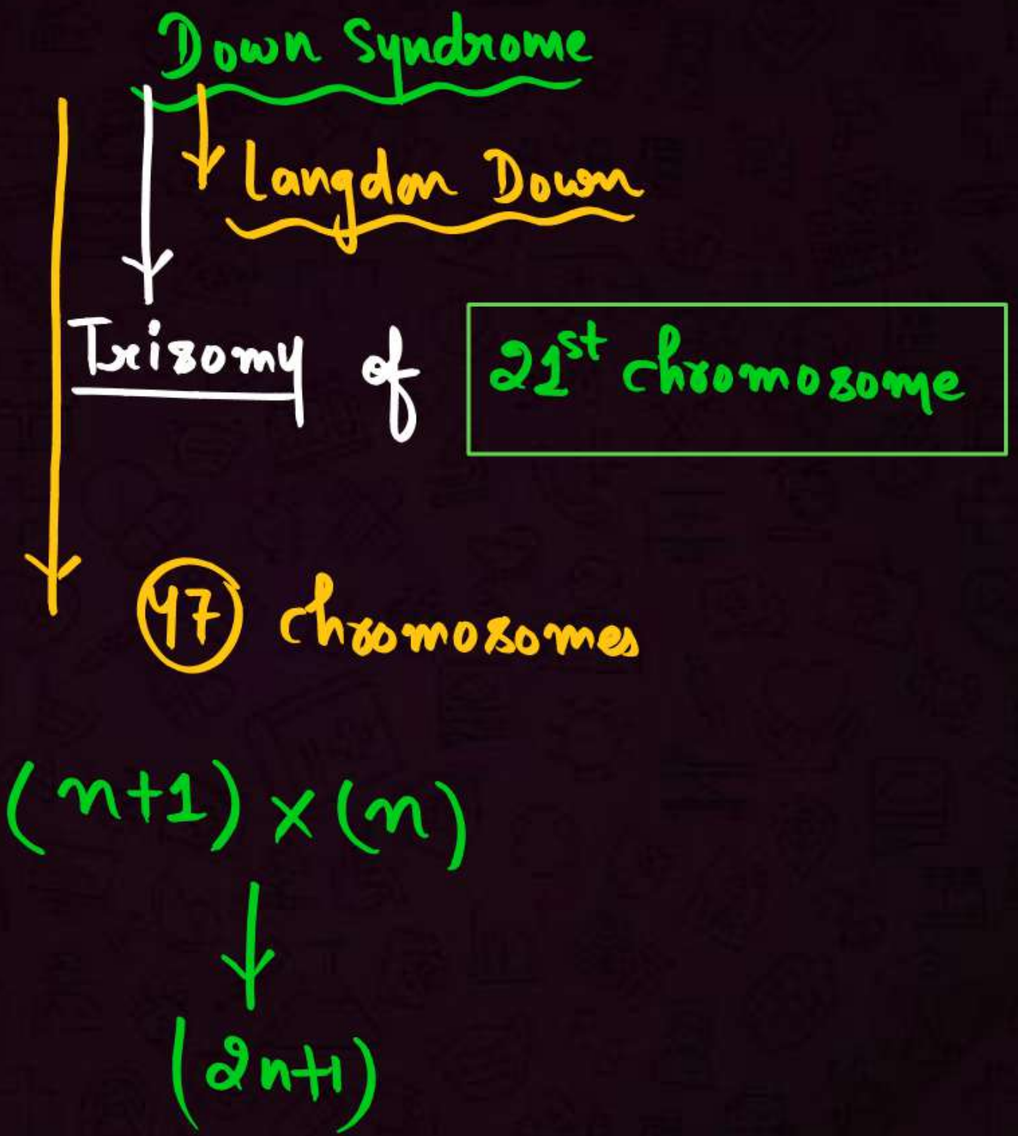
Autosomal

Dominant
- "Myotonic Muscular Dystrophy"

Recessive
- → Sickle-Cell Anaemia
- → Thallasaemia
- → PhenylKetonuria

X-linked

"Recessive"
- Color Blindness
- Haemophillia


Down Syndrome
Langdon Down
Trisomy of
21st chromosome
47 chromosomes
(n+1) × (n)
(2n+1)

Symptoms:

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

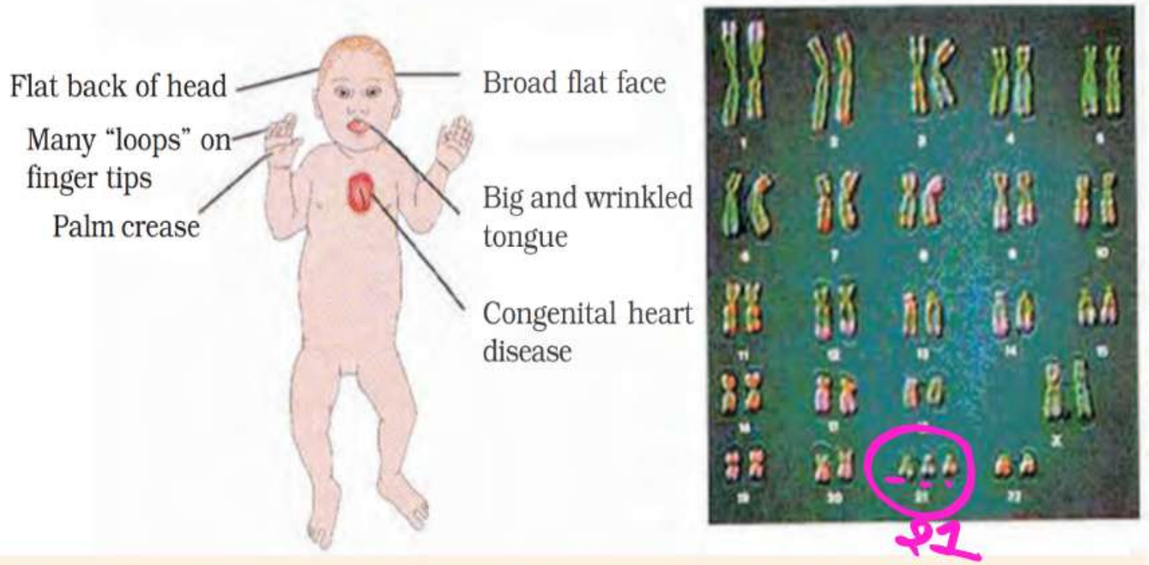


Figure 4.16 *A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual*

Klinefelter Syndrome

Normal male → 44 + XY

→ Trisomy of 23rd pair (Sex-chromosomes)

→ In Males → 44 + XXY → Extra X-chromosome in males.

or

47, XXY

Symptoms:

→ Gynaecomastia

↓

"Develop of Breasts"

(Feminine characters)

→ "Sterile"

Egg
Abnormal
22 + XX

× Normal sperm (22 + Y)

(22 + XY)

44 + XXY

Normal egg (22 + X) × Abnormal sperm (22 + XY)

↓

44 + XXY

Normal female
44+XX

Turner Syndrome

Monosomy of 23rd pair → (In females)

44+XO

(one X-chromosome)

or 45, XO

Symptoms:

a) Sterile
b) Short stature
c) Ovaries → Rudimentary
d) Secondary sexual characters not developed.

Abnormal gamete (n-1) × normal gamete (n)

↓

(2n-1)

PHENYLKETONURIA

Chromosome-12 → Gene → `PAH' gene → Phenylalanine hydroxylase Enzyme gene

Phenylalanine (amino acid) —"PAH" Enzyme→ Tyrosine (amino acid)

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

Phenylalanine (amino acid) ⟶✗ Tyrosine

↓

Accumulates as Phenyl pyruvic acid

↓

Mutated Gene

(Pleiotropic gene)

Phenylpyruvic acid accumulates in Brain cells → Mental Retardation

Excreted in Urine

→ Depigmentation in hair & skin

Thallasaemia → Needs Regular Blood Transfusion

α-Thallasaemia

- Only β-chains formed
- α-chain
  - Not formed (Major)
  - less formed (Minor)
- RBC → "Ruptures"
- "Cause" → Chromosome-16
  - HBA_1 Gene
  - HBA_2 Gene
  - (Total ④ alleles)
- "Short arm deletion"

β-Thallasaemia

- β-chains
  - Not formed
  - less formed

Cause: Chromosome-11 → HBB gene (mutation)

Sickle-Cell Anaemia

→ Point mutation

→ Substitution (Transversion)

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

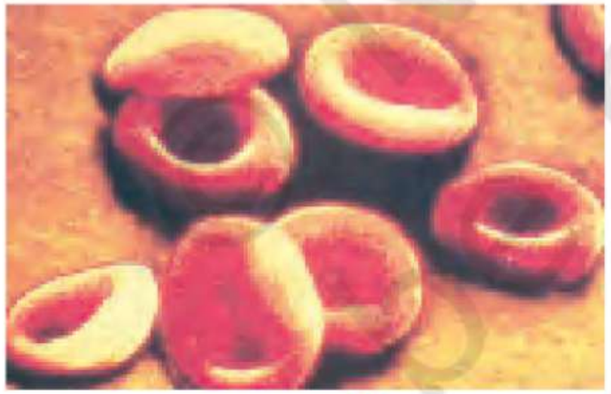
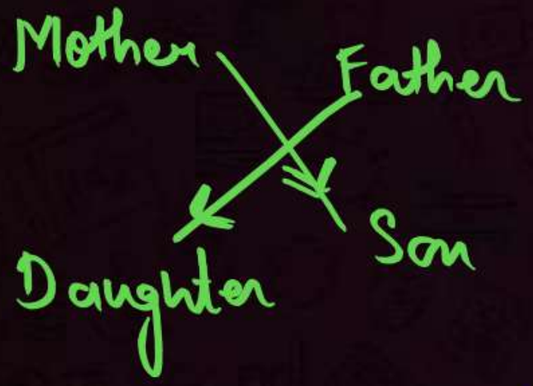


Figure 4.15 Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin: (a) From a normal individual; (b) From an individual with sickle-cell anaemia

Haemophillia

* * linked recessive disorder
* Bleeder's disease
* Coagulation factor (Any one of them)
  (Cascade effect)
  ↓
  Blood coagulation is Absent
* "Royal disease" (Pedigree of Queen Victoria)

Criss-Cross inheritance



XX → Normal
XX^h → Carrier
X^hX^h → Haemophillic

XY → Normal
X^hY → Haemophillic

Color-Blindness → X-linked recessive disorder

* Red-green color Blindness.
* Criss-cross inheritance
* Population
  - Males → 8 %
  - Females → 0.4 %

XX → Normal

XX^c → Carrier

X^cX^c → Color blind

XY → Normal

X^cY → Color Blind

* X-linked recessive disorders
  - Color Blindness
  - Haemophillia

are More common in Males than in Females

Reason:

Females

② recessive copies must be present

Males

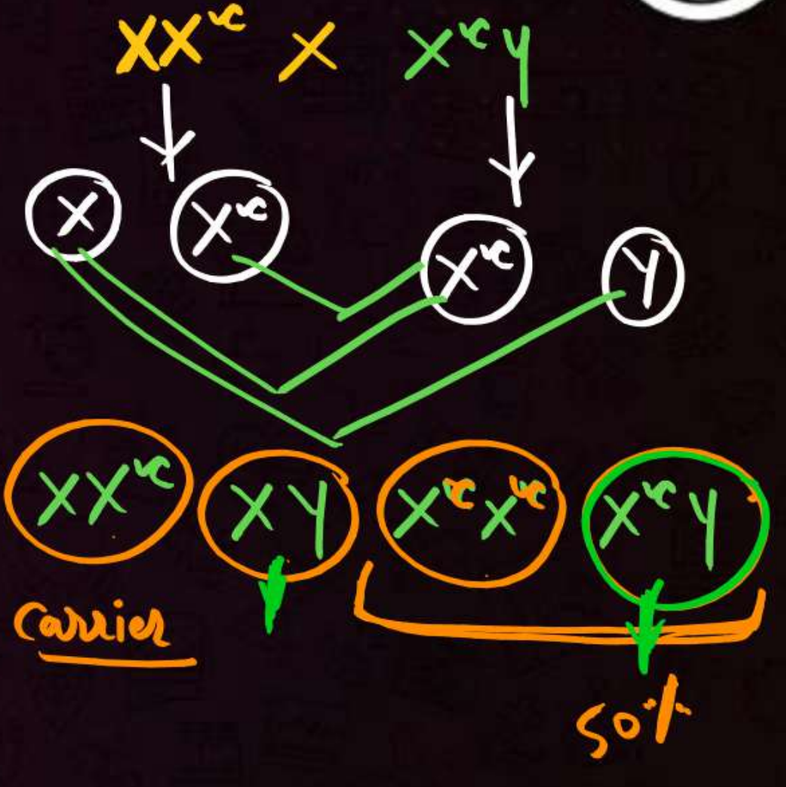
① recessive copy on X-chromosome is enough for disease.

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

a) what % of their progeny will be diseased → (50%)

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

c) what % of females will be color-blind → 50%



Pedigree-chart

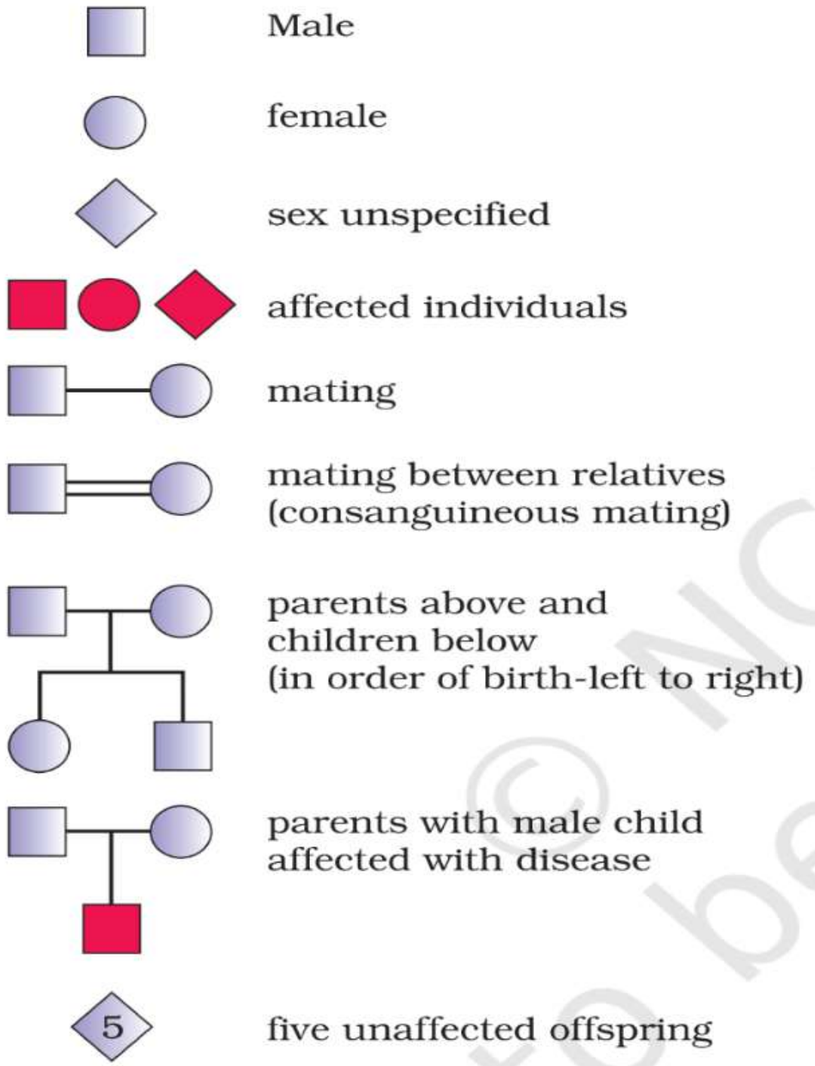
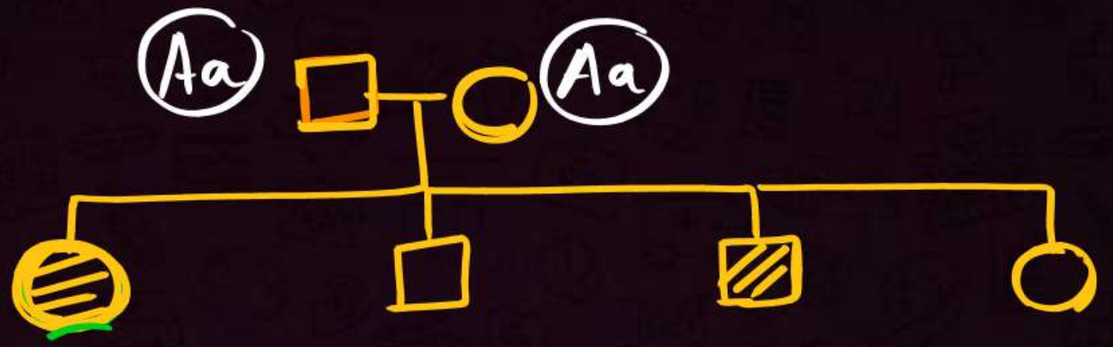


Figure 4.13 Symbols used in the human pedigree analysis

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



aa → Diseased

AA, Aa → Normal

Autosomal Recessive

Q 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

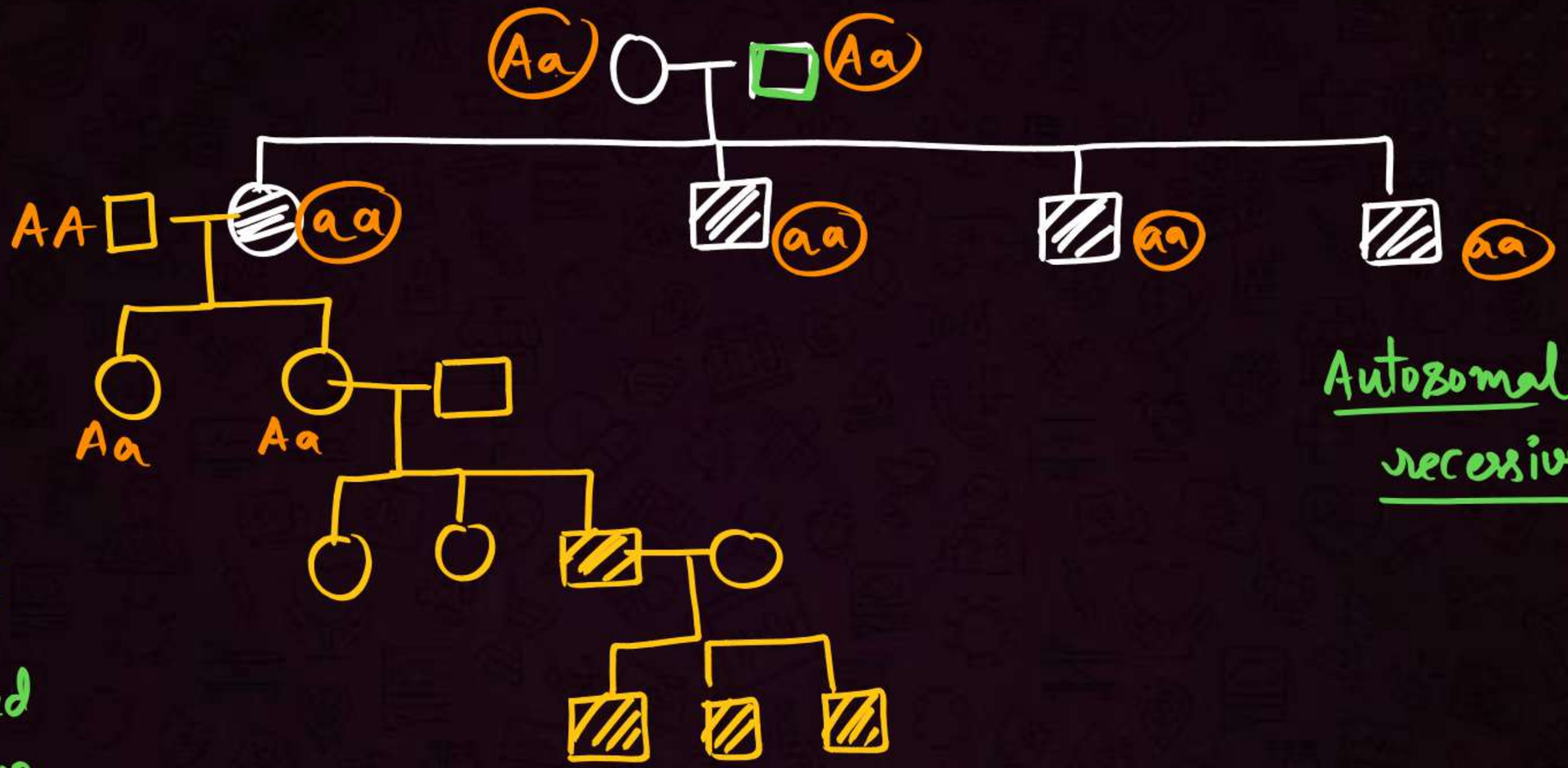
Cannot be **X-linked Recessive chart**

③ Mother → diseased
Son → Normal
→ Can't be X-linked recessive chart.

④ Mother → Normal
Son → Diseased

Cannot be X-linked Dominant chart

PYQ



~~Autosomal dominant~~

~~X-linked dominant~~

~~X-linked recessive~~

Autosomal recessive ✓

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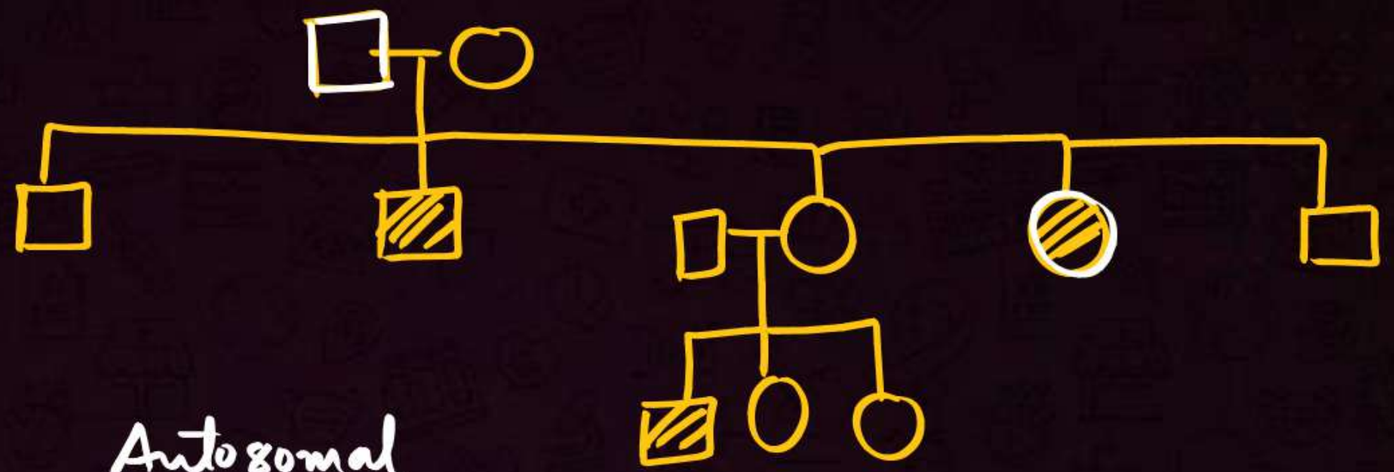
~~Dominant~~ chart

~~X-linked recessive~~



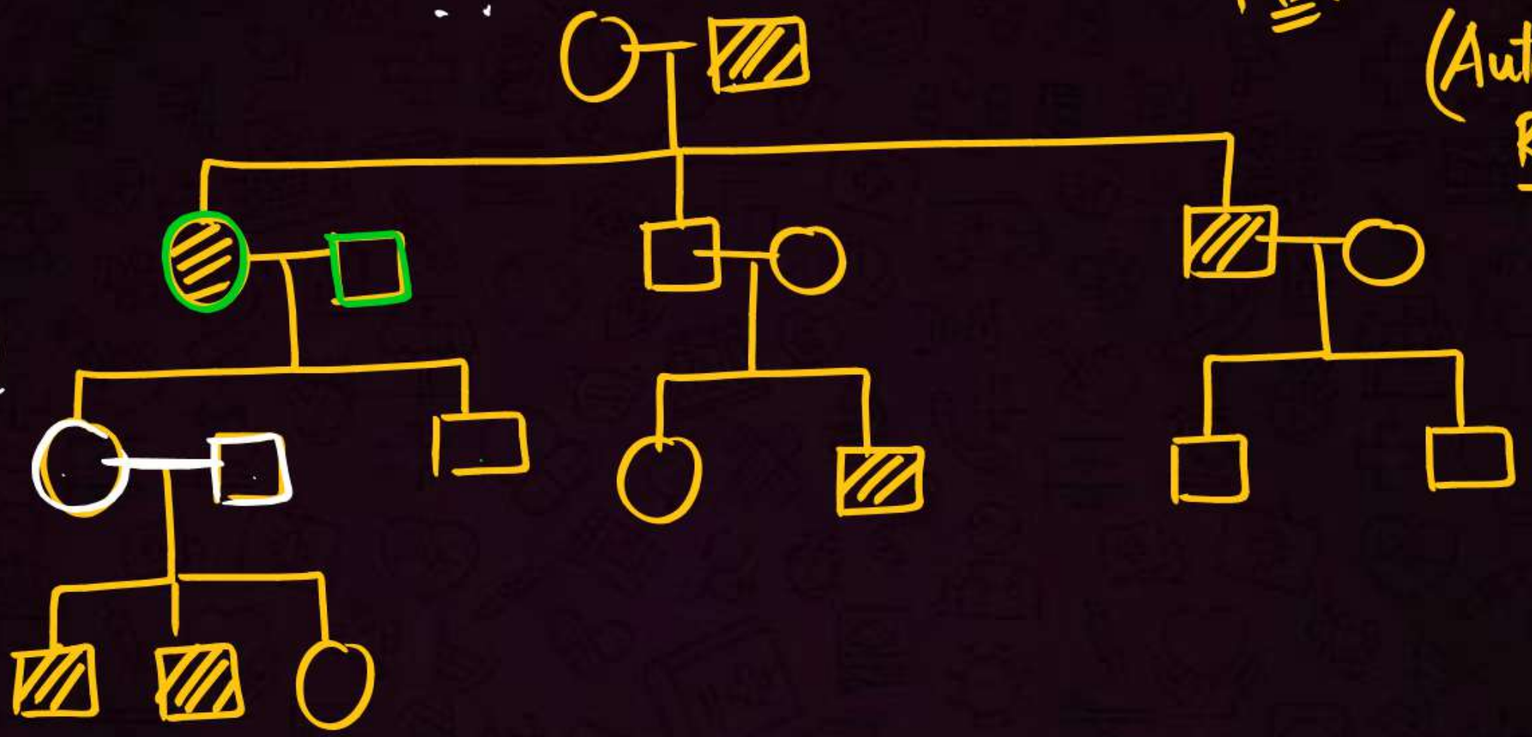
Autosomal recessive

Dominant chart

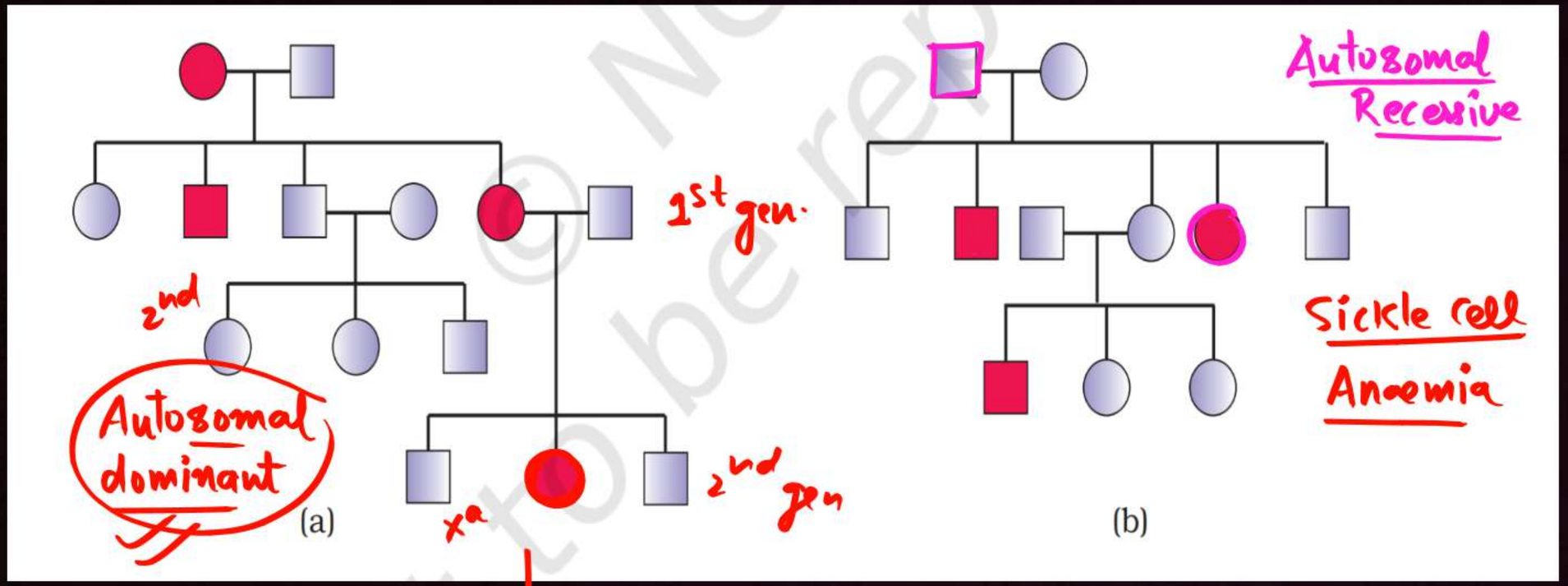
X-linked Autosomal

X-linked recessive

PYQ.



(Autosomal Recessive)



Myotonic Muscular Dystrophy

* Yakeen

↓

Uthaan series

↓

"Principles"

↓

"Questions"


THANK YOU