



Genetics

Concept of DNA, Chromatin & Chromosome

 DNA → Thread like str (Nucleic acid)
→ 2 Strand.
Macromolecule.

 Protein

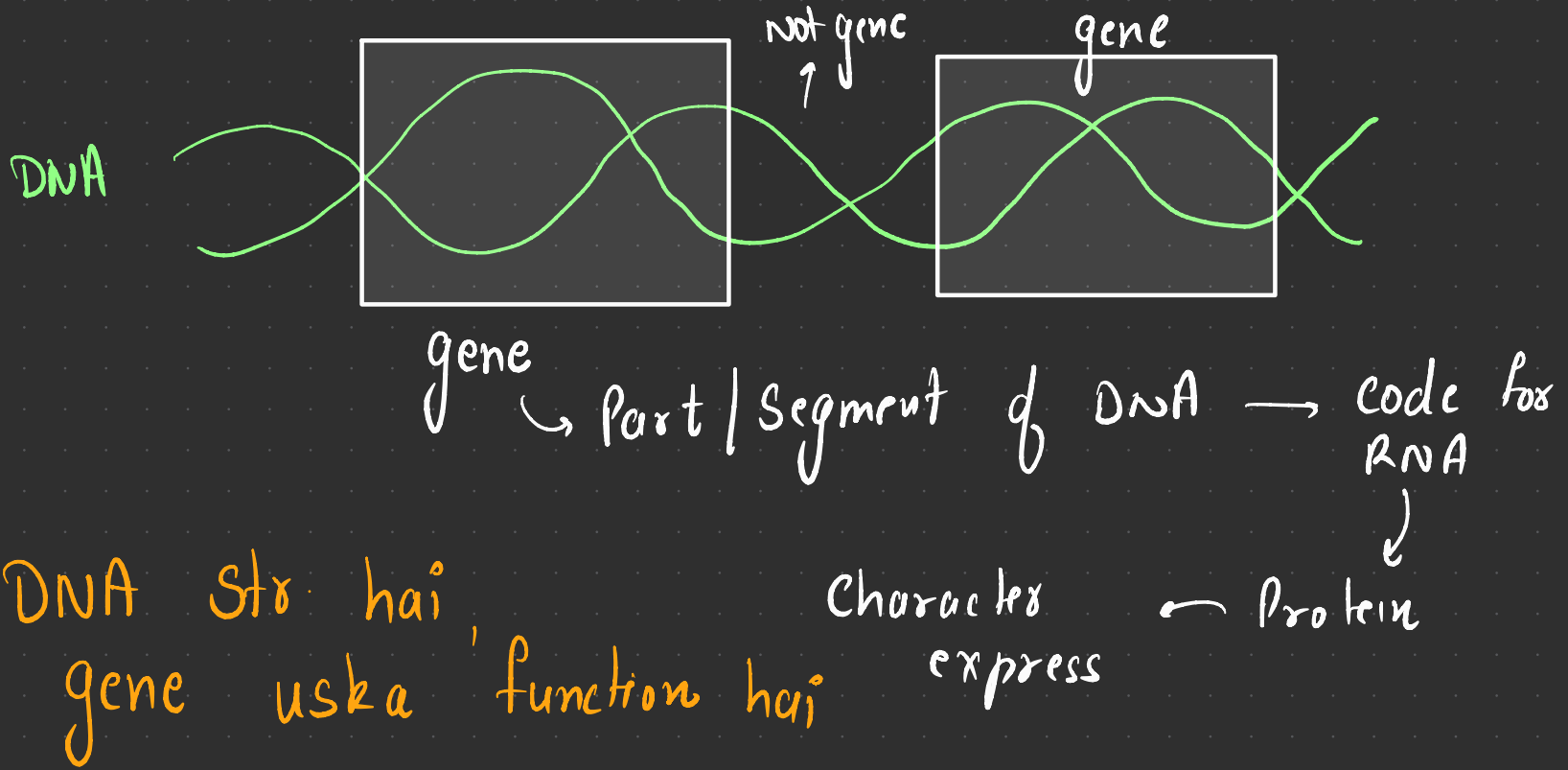
Chromatin

||
DNA

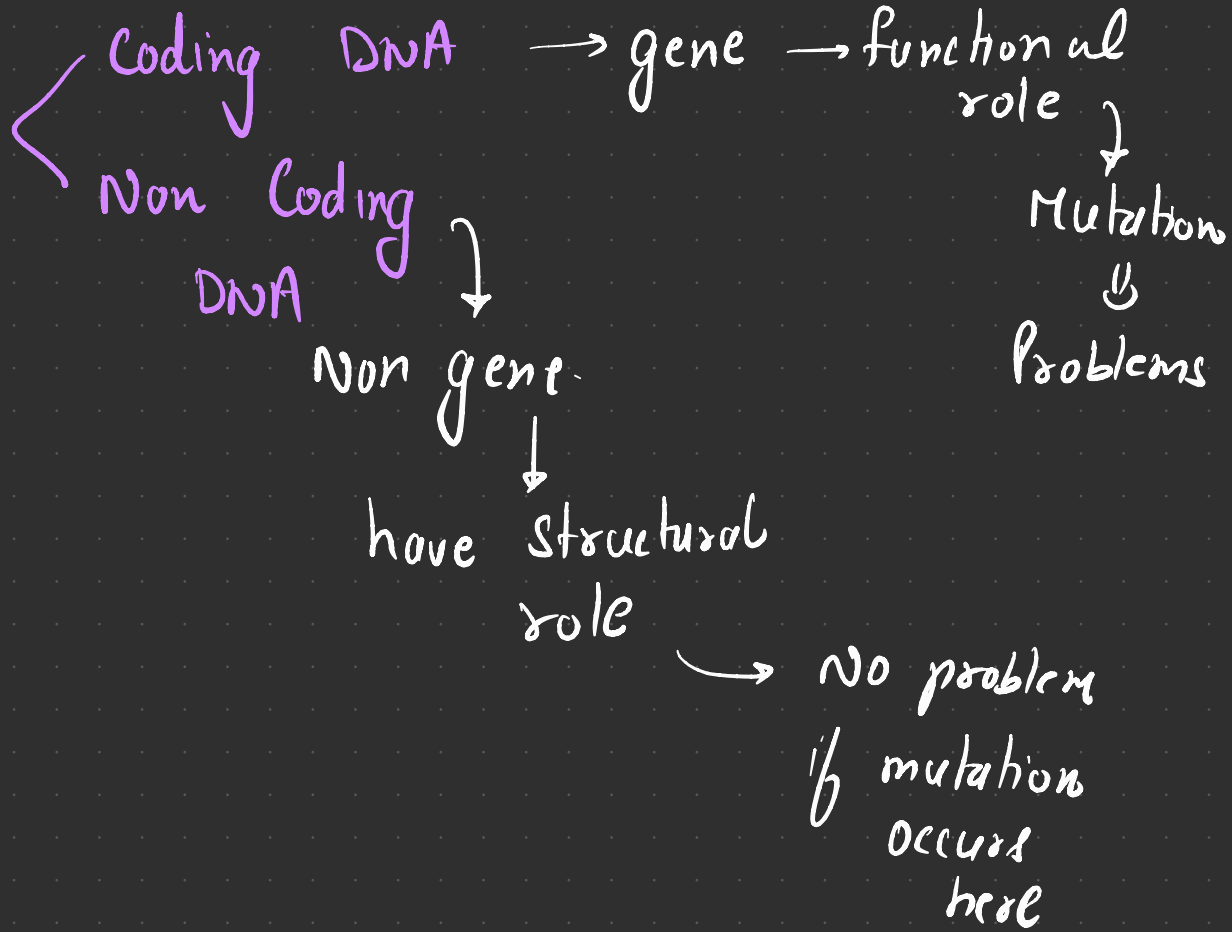
+
Proteins

 Chromosome

→ DNA + RNA
+
Protein



Total DNA
has
2 parts



Gene is Chemically DNA

$G_1 - S - G_2 \rightarrow$ Chromatin (Nucleus)

M phase $\rightarrow$ Chromosome (cytoplasm)

length of DNA $\rightarrow 2.2\text{ m} \rightarrow$ So condensed to Chromatin for storage

$\swarrow$
S phase

$\downarrow$
all DNA
get open
but not at once

$\downarrow$
DNA open in G_1 & G_2

$\downarrow$
for transport
(only gene part open)



DNA

Euchromatin

Chromatin

Heterochromatin

Chromosome

Condensatⁿ ↑
ability to make protein ↓

⇒ In most of cell → which are normally not dividing → Chromatin visible

⇒ dividing cell → Chromosome

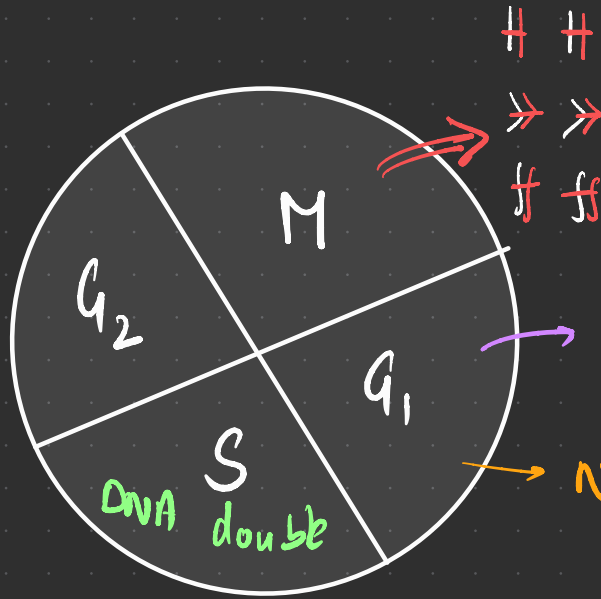
⇒ Chromatin of single cell → can form 46 chromosome
(single thread) $(2n = 46)$

lllll
lllll

|| ||

⇒ ⇒

ff ff

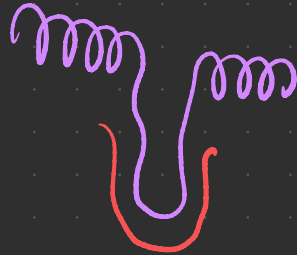


chromatin
lllll

No chromosome
visible

(if we draw)

| |
> >
s s



On chromatin, genes are randomly +nt, But when Chromosome form $\rightarrow$ Gene for 1 chr. always Comes at parallel location.

Concept of Character & trait

Eye Colour — Character $\left\{ \begin{array}{l} \text{Black} \\ \text{white} \end{array} \right\}$ Trait

Hair Colour — Character $\left\{ \begin{array}{l} \text{Blue} \\ \text{white} \end{array} \right\}$ Trait

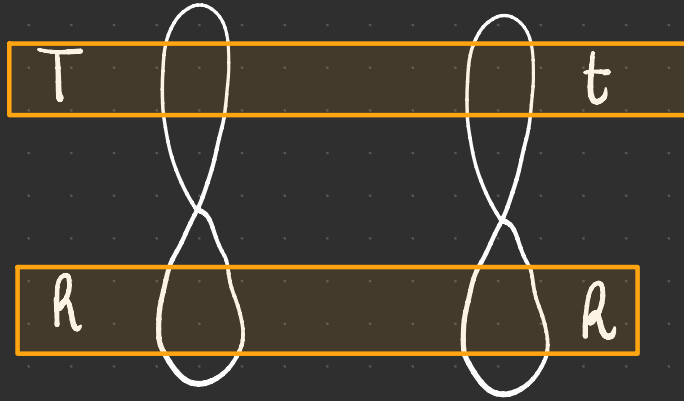
- * There are many character $\rightarrow$ Single Trait possible
- * for some character $\rightarrow$ 2 / more than 2 trait possible
- * Every chr. have fix position on chromosome
- ⊙ for any chr. single trait also possible

(eye position)

frontal $\leftarrow$ $\frac{1}{10}$ in 10 million

Mutation $\Leftarrow$ Back $\rightarrow$ 1 person

⇒ Concept of locus



→ locus (Heterozygous)

→ 1 for 1 chr.

locus (Homozygous)

1 locus = 2 gene (Diploid)
Some (y, y)
diff (T, t)

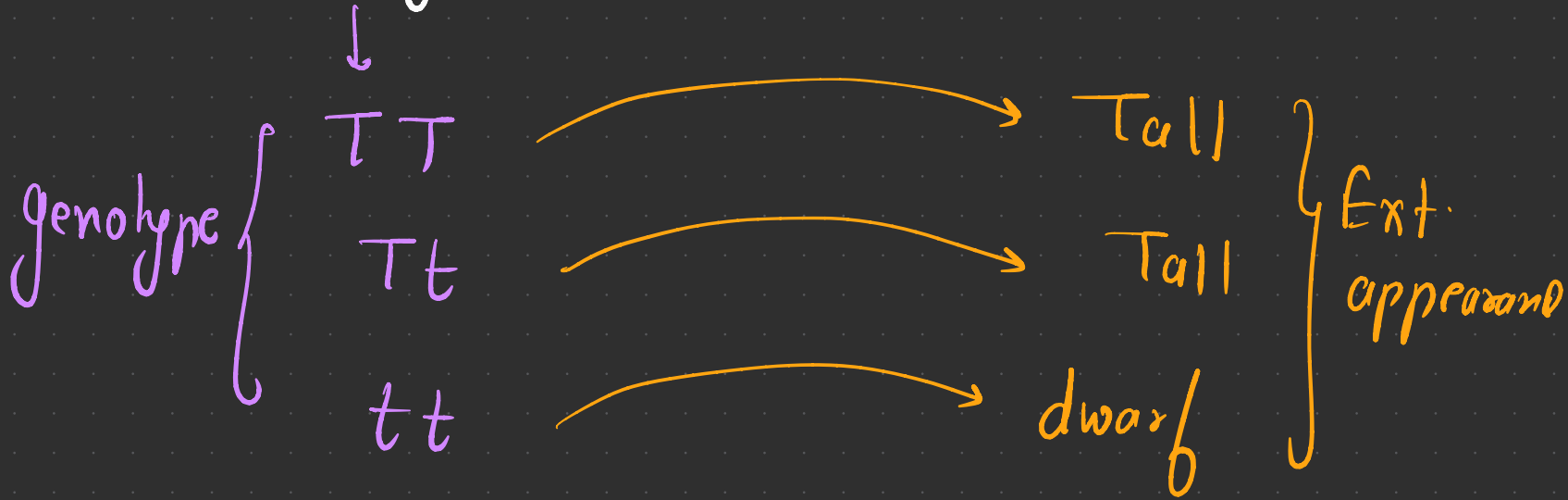


locus (Haploid) $\rightarrow$ Hemizygous

$\rightarrow$ 1 locus $\rightarrow$ 1 gene only

Genotype

Phenotype



TT AA - Diploid genotype

TA - Haploid genotype

TT - Diploid genotype

A - Haploid genotype

Genetics Branch of Bio deals with Heredity (transfer of trait) & variation

Heredity $\Rightarrow$ Transfer of Chr parents $\rightarrow$ offspring

Variation Amt / degree of diff. b/w
offspring & parent

Process
" inheritance

Continuous discont.

Source of variation $\rightarrow$ Sexual Repro
 $\rightarrow$ Mutation

Evolution of Genetics

① 800 - 1000 BC → Agriculture — Human know
Evolve
↓
Source of
Variatⁿ in sexual
Repro

② Pre mendelion Concept → Scientist worked on genetics before
medal
- Based on Blending inheritance
(Mixing of Trait)

Details

Exp. years - 1856-63

Published - 1865

Experimented on

→ *Pisum sativum*

→ 14 breeding
variety

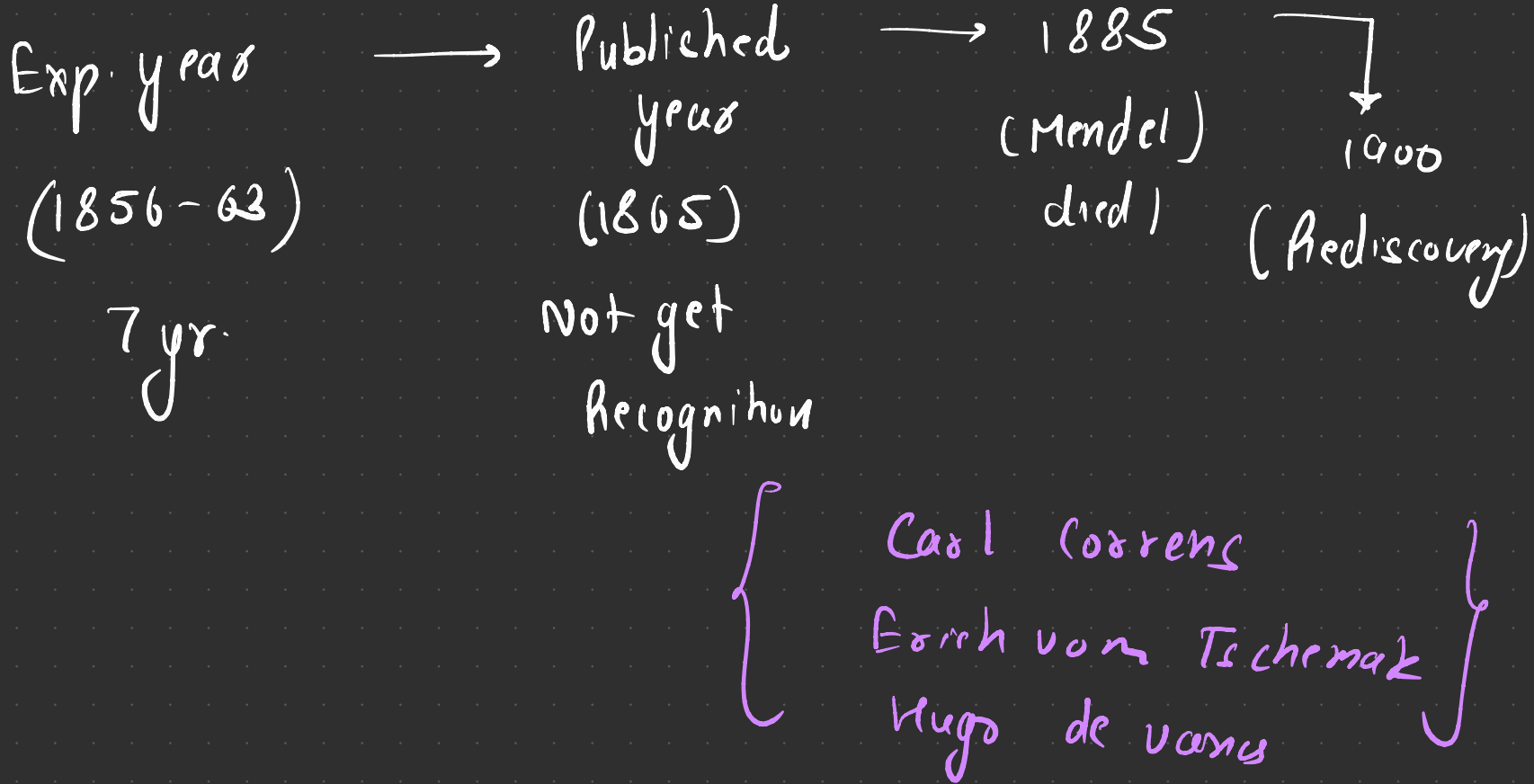
Why pea → can be easily grown

→ self pollinat^y but can
be cross pollinated

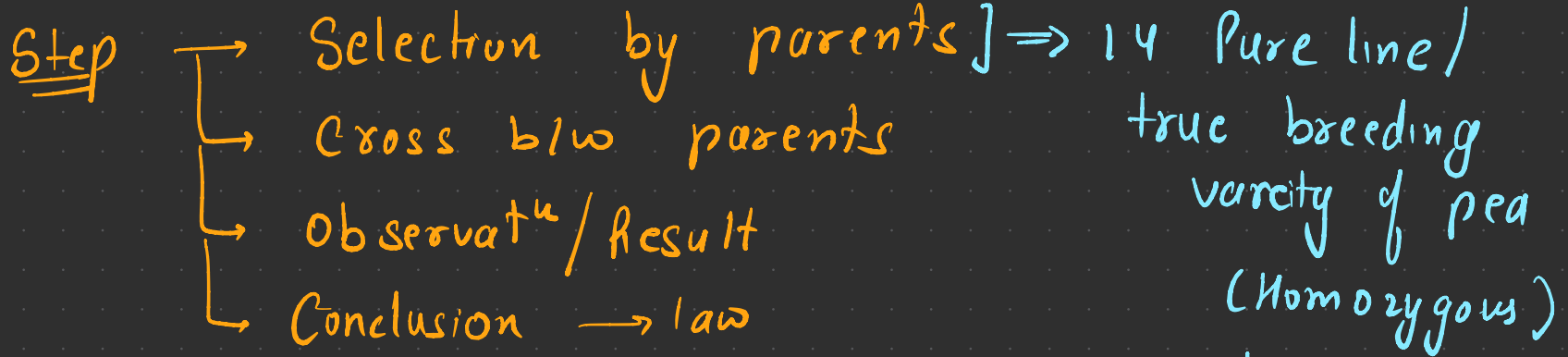
+ chr. easily observable

→ complete life cycle → in single
season

Rediscover by Scientist



Hybridization Experiment → by Mendel



Pea $2n=14$

↓
Repeated selfing

↓
 $\begin{matrix} TT \\ tt \end{matrix} \rightarrow \text{True breeding variety}$

$Tt \rightarrow \text{Not true breeding}$

- 14 true breeding line / Pure $\rightarrow$ differ in single trait
- tall $\longleftrightarrow$ dwarf.

γ Characters $\rightarrow$ Trait

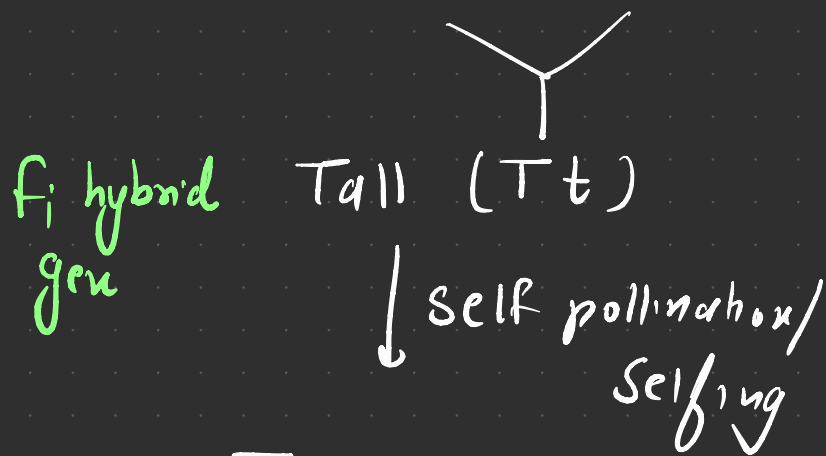
		(D)	(R)
Plant height		Tall	dwarf
flowers	colour	Violet	white
	Position	Axial	terminal
Pod	Shape	Smooth	Constricted
	colour	Green	yellow
Seed	Shape	Round	wrinkled
	colour	Yellow	Green

② Cross b/w Parents

Pure parents (cross)



Pure tall × Pure dwarf
TT tt



F₂ gen ← Tall, dwarf

③ Observation / Result

Tall $\times$ dwarf



(F₁) Tall



(F₂) Tall Tall Tall dwarf $\Rightarrow$ 3 tall
1 dwarf

$\rightarrow$ F₁ hybrid is exactly similar to any of parent

$\rightarrow$ F₂ match / similar to both parent

$\rightarrow$ Any one (tall) is more in no. than other progeny (3:1)

④ Conclusion

Something being stably passed in unchanged manner from one gen to next. which control trait

↓
factor !! (now klas gene)

factor

- occur in pairs
- Control trait
- Not show Blending / Mixing
- from parent → offspring via gamete

Mendel Law

1) Law of inheritance

(a) law of dominance $\longrightarrow$ Trait controlled by discrete unit c/d factor

$\longrightarrow$ factor occur in pair

$\longrightarrow$ In dissimilar pair (Tt) $\longrightarrow$ Show $\downarrow$ dominance

$\longrightarrow$ Req. F_1 & F_2 for proof

(b) law of Segregation

→ gamete formation → factor segregates
50%

Chance of any of factors

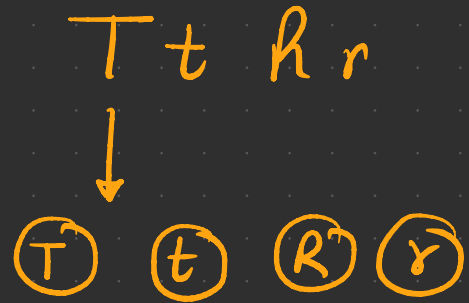
→ Factor not show blending.

Q A plant of genotype $TtRr$ produce pollen
↓
% of pollen

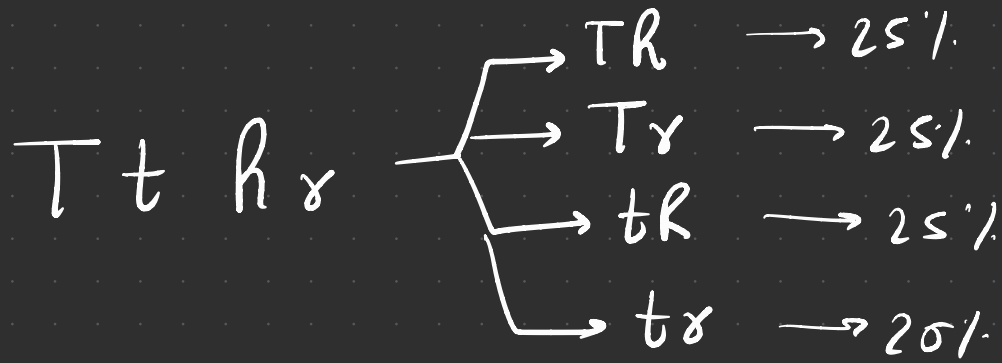
a) TR gene $\Rightarrow 25\%$

b) tR gene $\Rightarrow 25\%$

c) tR gene $\Rightarrow 50\%$



$$\frac{1}{2} \times \frac{1}{2} = \frac{1}{4} \left(\begin{array}{l} \frac{1}{2} \text{ pollen } (T) \\ \frac{1}{2} \text{ pollen } (R) \end{array} \right) \downarrow 25\%$$



Q. Plant - Pea - Genotype $\rightarrow \overline{Tt} \overline{Rr} \overline{Yy}$

% of pollen $\Rightarrow \overline{TRY}$
 $\downarrow \downarrow \searrow$
 $\frac{1}{2} \frac{1}{2} \frac{1}{2} = \frac{1}{8} = 12.5\%$

% of pollen $\overline{TY}$
 $\downarrow \downarrow$
 $\frac{1}{2} \frac{1}{2} = \frac{1}{4} = 25\%$

of 1000 pollen
 $\downarrow$
 125 (TRY)

$$\underline{Q} \quad \underline{TT} \quad Rr \quad Yy$$

$\frac{1}{2} \quad \frac{1}{2} \quad \frac{1}{2}$

% of Pollen

$$- \underline{TR} \underline{Y}$$

$1 \quad \frac{1}{2} \quad \frac{1}{2} = \frac{1}{4} = 25\%$

Why Mendel work was special:-

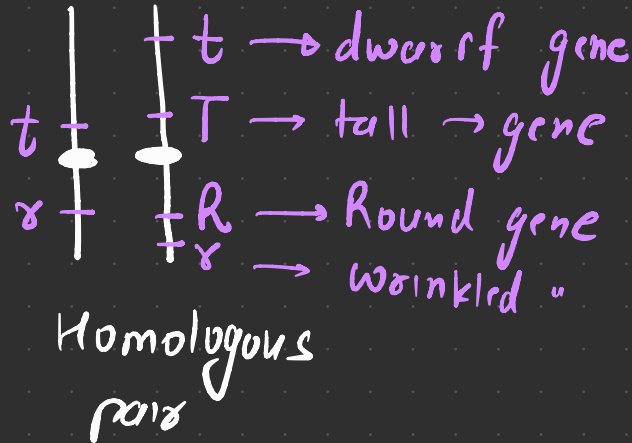
- use Maths logic & Statistical analysis → for his cross
- Large sampling size
- Repeated selfing - Result check
- Mendel proposed general principle rather un substantiated data.

Concept of gene & allele

1 gene have 2 allele X
1 allele have 2 gene X

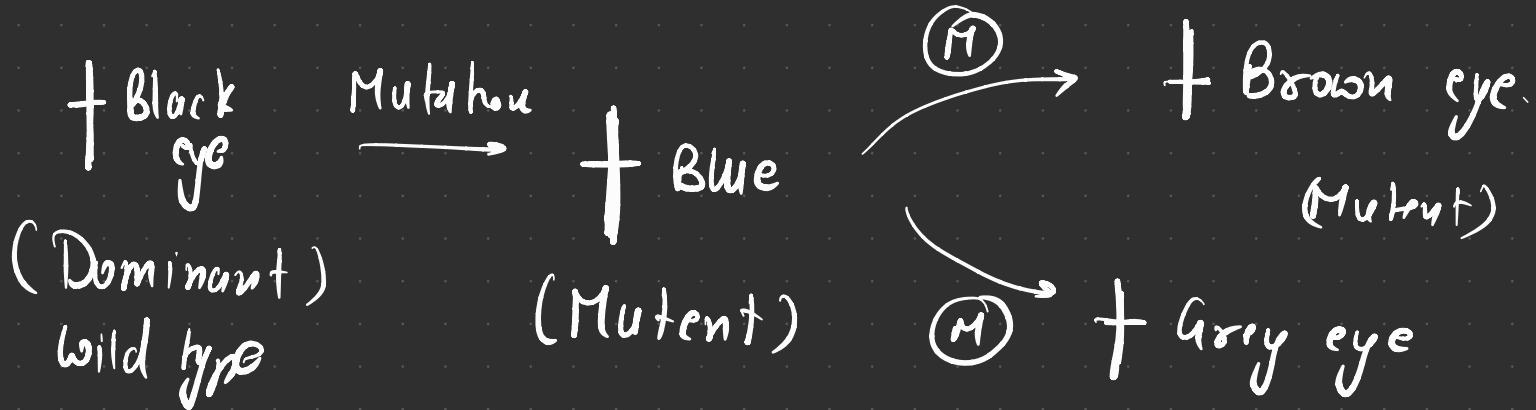
} Allele are those gene which have info for contrasting trait

R, r → Related



{ T, t - Related
(they have same locus, they belong to same chr.
(Plant height))

for 1 Chr $\rightarrow$ 1 allele $\rightarrow$ Possible (Eye position) $\downarrow$
for 1 Chr $\rightarrow$ 1 allele $\rightarrow$ Possible $\downarrow$ frontal (colour)
Eye colour $\leftarrow$ Black (allele)
Blue (allele)



Gene - feature express

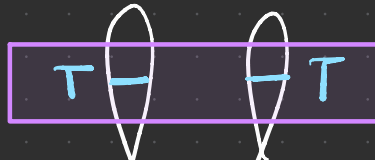
Allele $\rightarrow$ word to show relation

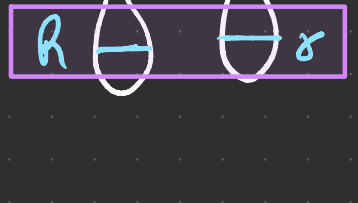
Trait $\rightarrow$ feature

Gene/allele which evolve 1st for any chr. $\rightarrow$ wild dominant

Single individual $\rightarrow$ 1 Chromosome pair $\rightarrow$ Max^m 2 allele

Population $\rightarrow$ 1 chr. $\rightarrow$ Multiple allele.

 $\Rightarrow$ 2 allele = similar type $\rightarrow$ Homozygous

 $\Rightarrow$ 2 allele = Contrary
 or
 dissimilar $\rightarrow$ Heterozygous.

Multiple allele

→ In a population → 1 chr

→ 2 gene.
(Minimum
2 individual)

Eye colour
↓
Allele

(3)

Black
eye

Blue
eye

Brown
eye

diff. in
individual

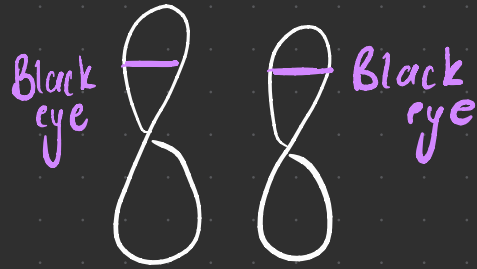
Same

locus.

Chromosome
no

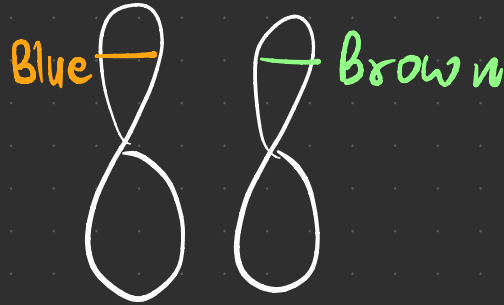
Chr

Human x



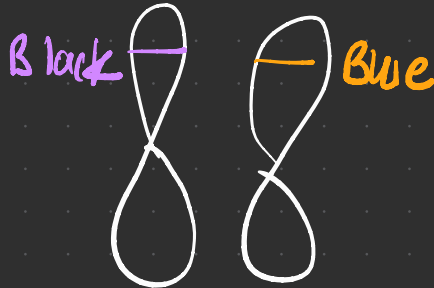
→ Homozygous

Human y



→ Heterozygous

Human z



→ Heterozygous

Monohybrid cross

Single character consider / Single locus

Pure TT (tall) $\times$ tt dwarf } Parental gen

$\downarrow$
 T

$\downarrow$
 t

Tt Tall

} F_1 gene

$\downarrow$ selfing

TT
Tall

Tt
Tall

Tt
Tall

tt
dwarf

} F_2 gen

⑧ Mendel not drawn any punnet square

↳ He punnet

(British

↳ Graphical representation
to show possible
genotype

Tall x dwarf (Parental)



Phenotype $\leftarrow$ Tall $\leftarrow F_1$

genotype $\leftarrow$ Tt



		T	t
T	T	TT	Tt
	t	Tt	tt

Phenotype $\leftarrow$ 3 tall 1 dwarf
 genotype $\leftarrow$ TT Tt Tt tt

(F_2)

No. of phenotype

= 2

$\leftarrow$ tall
 $\leftarrow$ dwarf

$\rightarrow 3/4$

$\rightarrow 1/4$

No. of genotype

= 3

$\leftarrow$ TT $\rightarrow 1/4$
 $\leftarrow$ Tt $\rightarrow 2/4$
 $\leftarrow$ tt $\rightarrow 1/4$

Phenotype
ratio

Tall : dwarf

$3 : 1$

Genotypic
ratio

TT

Tt

tt

1

:

2

:

1

Dihybrid cross

2 gene locus consid.

Tall Round
(TT RR)

x dwarf wrinkled } Parental
(tt rr)

Gametes $\downarrow$
TR



$Tt Rr$

$\downarrow$ tr Gametes

$F_1 \rightarrow \textcircled{P} = 1$
 $\rightarrow \textcircled{Q} = 1$

$\downarrow$ selfing

$\textcircled{P} \quad 9 : 3 : 3 : 1$

$\textcircled{Q} \quad 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1$

$TtRr$



$(T) < \begin{matrix} R \\ r \end{matrix}$

$\rightarrow TR$

$\rightarrow Tr$

$(t) < \begin{matrix} R \\ r \end{matrix}$

$\rightarrow tR$

$\rightarrow tr$

} 4 Gametes ✓

$\frac{R}{r}$	TR	T γ	t γ	tR
TR	TTRR	TTR γ	Tt R γ	Tt RR
T γ	TTR γ	TT $\gamma\gamma$	Tt $\gamma\gamma$	Tt R γ
t γ	Tt R γ	Tt $\gamma\gamma$	tt $\gamma\gamma$	tt R γ
tR	Tt RR	Tt R γ	tt R γ	tt RR

phenotype $\Rightarrow$ ④

$$\text{Tall Round} = \frac{9}{16}$$

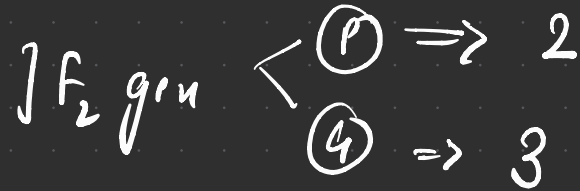
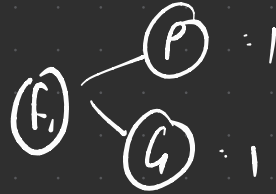
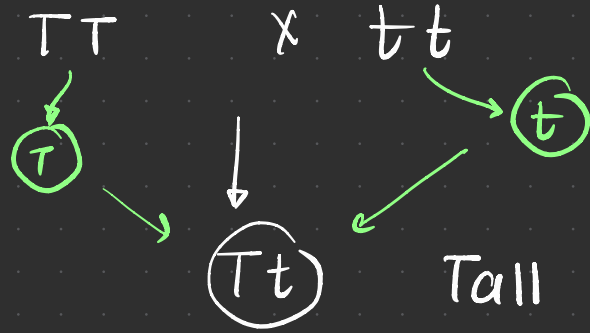
$$\text{Tall wrinkled} = \frac{3}{16}$$

$$\text{dwarf Round} = \frac{3}{16}$$

$$\text{dwarf wrinkled} = \frac{1}{16}$$

Genotype = 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

Monohybrid



Dihybrid cross

Tall Round

$TT RR$



x Dwarf wrinkled

$tt rr$



Multiply 2 monohybrid crosses



Dihybrid cross result

$Tt Rr$ F_1 gene

Tall Round



④

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

⑤

$9 : 3 : 3 : 1$

} F_2

Trihybrid cross

Tall Round Yellow

$TT RR yy$

x dwarf

wrinkled green

$tt rr yy$

$Tt Rr Yy$

↓ ⑤

①

Tt

Rr

Yy

x

Tt

Rr

Yy

Tall = $\frac{3}{4}$

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

Round = $\frac{3}{4}$

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

Yellow = $\frac{3}{4}$

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

tall

- Round yellow = $\frac{27}{64}$
- Round green = $\frac{9}{64}$
- wrinkled yellow = $\frac{9}{64}$
- wrinkled green = $\frac{3}{64}$

dwarf

- Round yellow = $\frac{9}{64}$
- Round green = $\frac{3}{64}$
- wrinkled yellow = $\frac{3}{64}$
- wrinkled green = $\frac{1}{64}$

⑦ Total Gamete $2^n \rightarrow$ no. of Heterozygous.

$$P = 2^n$$

$$G = 3^n$$

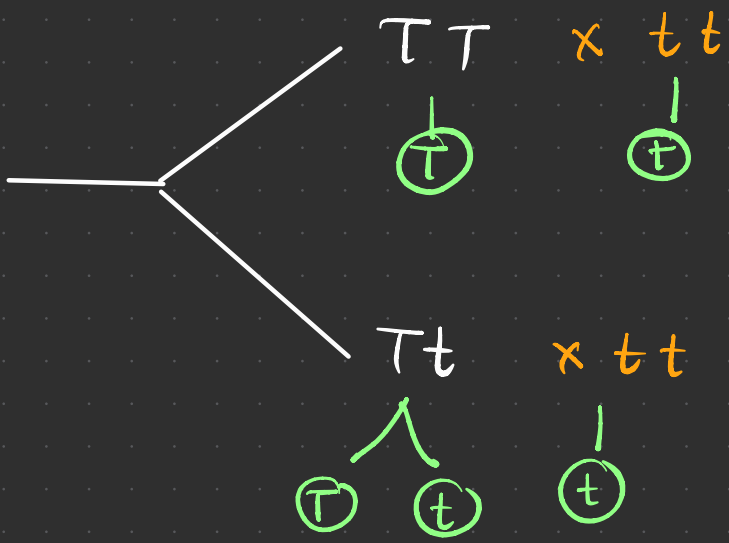
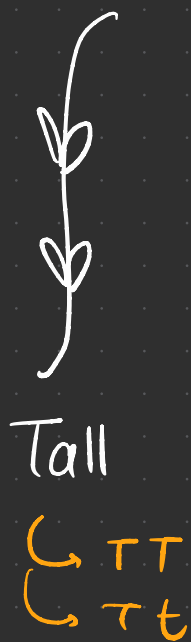
$$\text{Progeny / Box of Punnet} = 4^n$$

Test cross

- * To find unknown genotype
- * To find type of gamete

→ cross made with unknown (Q) x Recessive parent

→ Always $1:1 = (P) : (Q)$



→ All tall progeny

→ Tt Tt

tt tt

tall $2 \frac{1}{2}$

dwarf $2 \frac{1}{2}$



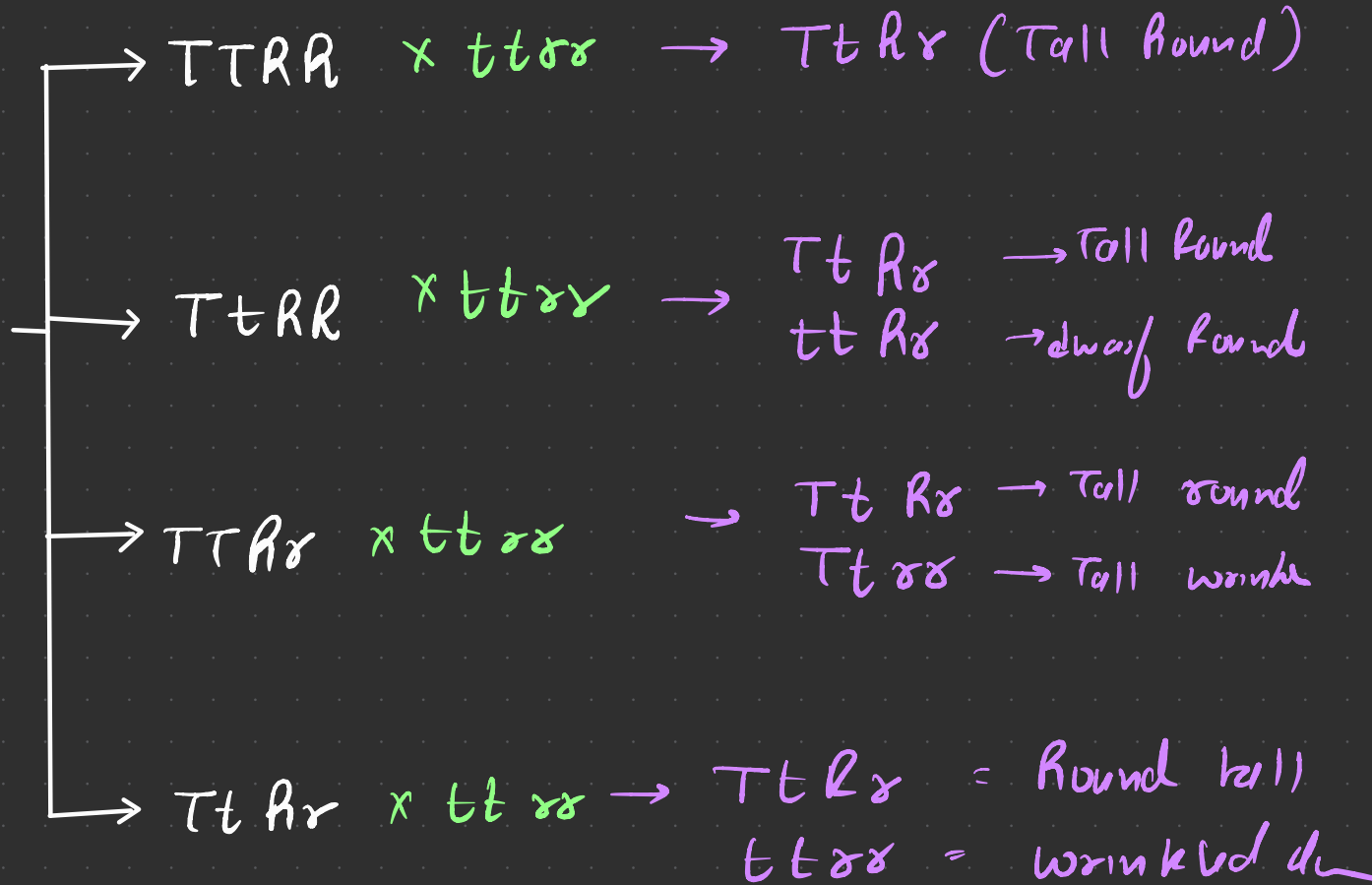
Unknown is

Heterozygous tall

Dihybrid Test cross

100%

Tall Round
(4) Possible
Genotype



Q Give Phenotype

a) 2

~~b) 4~~

c) 3

d) 1

TtRr

x

Ttss

(T) (t) (R) (r)

(T) (t) (s) (s)

(4) {
TR
Ts
tR
tr

(2) {
Ts
tr

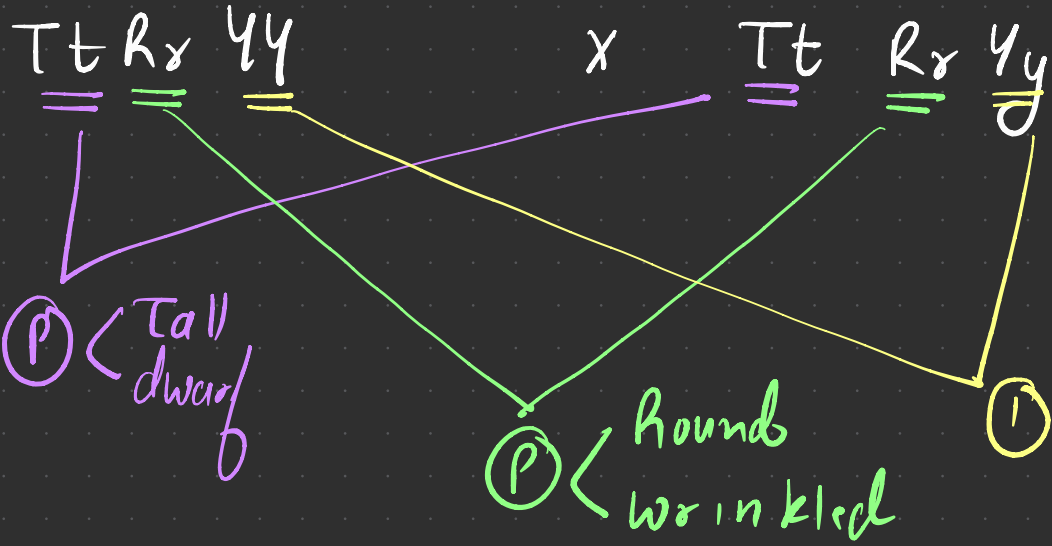
(4) {
Tall Round
Tall wrinkled
dwarf "
dwarf Round

Q No. of gamete $\rightarrow$ $Aa Bb$
 $\rightarrow$ $Rr Yy$

$$a) Aa Bb = 2^n = 2^2 = 4 \begin{array}{|l} AB \\ Ab \\ aB \\ ab \end{array}$$

$$b) Rr Yy = 2^n = 2^1 = 2 \begin{array}{|l} RY \\ Ry \end{array}$$

①



$P = 2 \times 2 \times 1 = 4$

Back cross

cross with any 1 parent

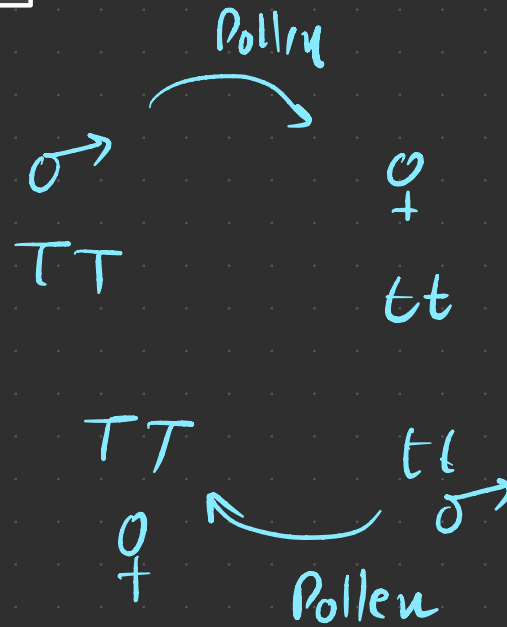
$TT \times tt$

Tt

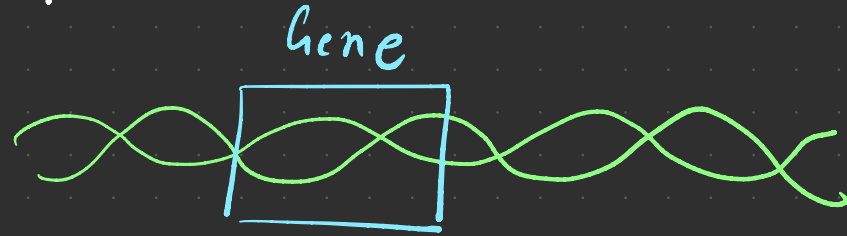
$TT \quad Tt \quad Tt \quad tt$

use $\downarrow$
Homozygosity $\uparrow$

Reciprocal Cross



How gene express



5' ————— 3' mRNA



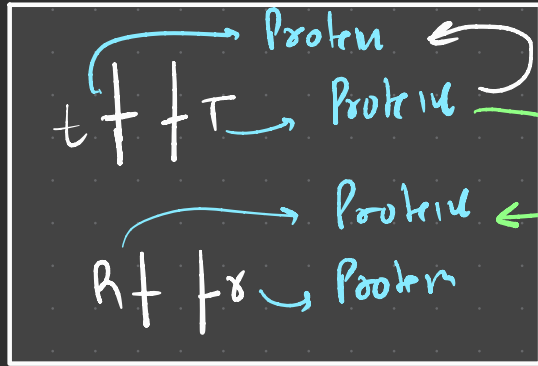
Protein



Trait Express

Gene interaction

In gene interaction, it is protein which interact



interaction of 2 gene of 1 locus
or
Allele within one locus.

Interaction of 2 gene / allele of
diff locus] b/w 2
locus

Single gene loci

T & t
interact

(Monohybrid)

2 gene loci

T & R
interact

(Dihybrid)

eg ① Dominance. $\downarrow$ acc. to Mendel

Not follow
Mendel's
rule, { Incomplete dominance
Co-dominance
Pleiotropy
Multiple alleles

Dominance

Red white (P)



all Red

(F₁)

→ F₁ will match any of one parent



R

R

r

w

(F₂)

Incomplete dominance

red

white

(P)

↓
Pink

F_1

→

F_1 will not
match with any
of the parent

↓ (S)

R

R

R

w

→ (F_2)

Codominance

red

white



(F₁)

→ F₁ will match
with both
parents



(S)

h



w

(F₂)

Incomplete dominance

$$(P) = (G) = 1:2:1$$

- * F_1 will not resemble any of the 2 parent
- F_1 will be intermediate of both.

Red flower RR X white flower rr (Parental)

↓
Pink flower Rr

↓ Selfing

RR Rr Rr rr
Red Pink Pink White

Flower colour → *Mirabilis Jalapa*
(4 'o clock plant)

Flowers - Snapdragon, dog flowers,

Antirrhinum majus.

→ genotype $\begin{matrix} RR \\ Rr \\ rr \end{matrix}$ $\frac{1}{2}$
→ phenotype $\begin{matrix} \text{Red} \\ \text{Pink} \\ \text{White} \end{matrix}$ $\frac{1}{2}$

Co-dominance

- * F₁ gen resemble both parent completely
- * Both gene not dominant
- * Both express themselves in F₁ gen

eg ABO sys → AB Blood grp

$Hb^A Hb^S$ - Carrier in sickle cell Anaemia

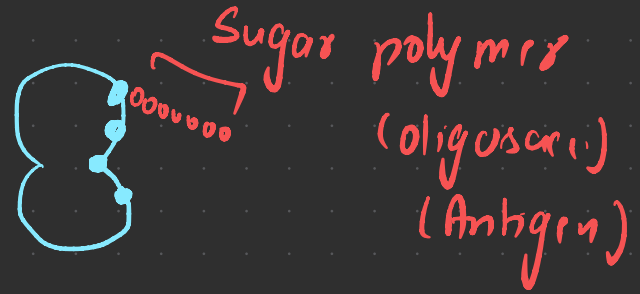
Symbol for gene · If any character - tall, dwarf (Tt)

If character more than 2 trait ⇒ Blood grp $I^A I^B i$

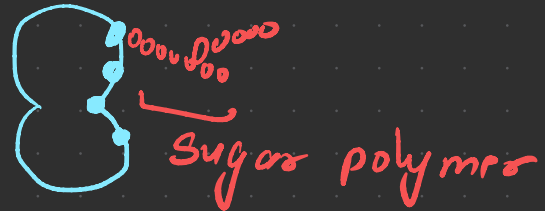
ABO Blood group

In human Gene I - Control blood group $\rightarrow$ 3 allele - I^A
 I^B

$I^A \rightarrow$ A antigen on RBC
 $\downarrow$
due to specific
sugar polymer



$I^B \rightarrow$ code for B antigen on RBC



i $\rightarrow$ not code for any antigen

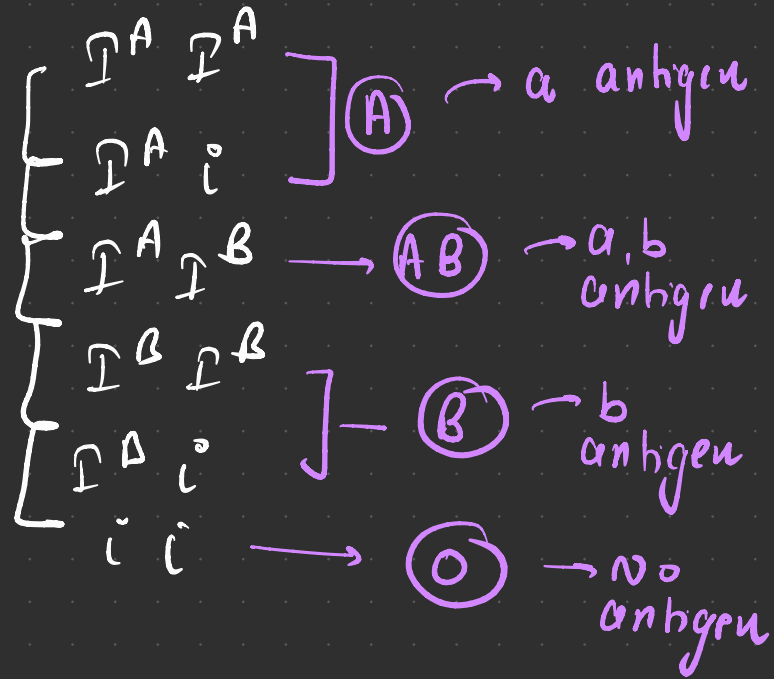


I^A

I^B

i

Max^m
6 combination
possible



→ 3 allele (in population) (Multiple allele)

→ 4 phenotype → A , AB , B , O

6 Genotype

→ $I^A I^A$ $I^A I^A$ $I^B i$ $i i$
 $I^A i$ $I^B I^B$

Blood group

is

example of

→ Multiple allele

→ dominance ($I^A i$) = A

→ Codominance ($I^A I^B$)

Sickle Cell anaemia



Round
Biconcave

$Hb^A Hb^A$



Sickle
Shape

$Hb^S Hb^S$

normal



$Hb^A Hb^S$

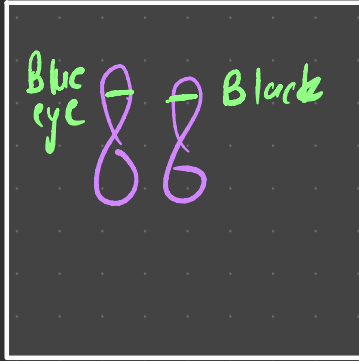
$O_2 \downarrow$

Some RBC
become
sickle
shape
 $Hb^A Hb^S$

Multiple allele

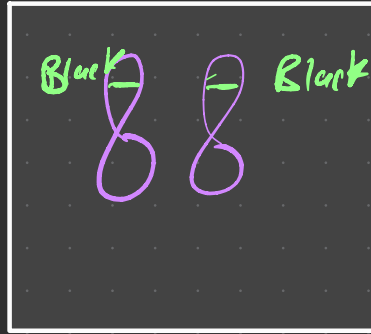
- Same locus / character / chromosome no.
- More than 1 individual

CH-10



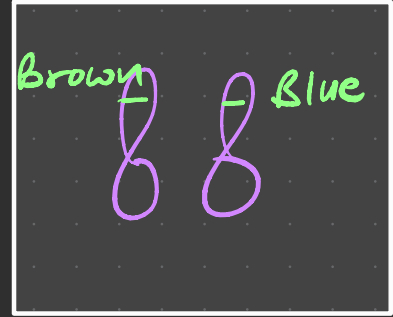
Ram

CH-10



Shyam

CH-10



Anishka

1 human $\max^m = 2$ allele.

Blood group (3 allele $\rightarrow$ 6 $\textcircled{G}$)

Drosophilla eye colour (15 allele)

$\rightarrow$ Evolve due to mutation on same locus & selection in nature

$\rightarrow$ first evolve $\rightarrow$ wild type $\rightarrow$ dominant

$\rightarrow$ we cannot predict $\textcircled{P}$ in case of Multiple allele

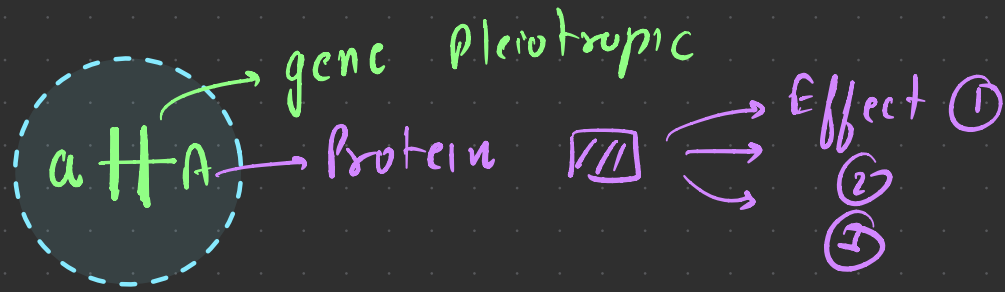
$\downarrow$
Mutant allele can show
dominance, codom, Incomp. dom

→ Mutant recessive over wild

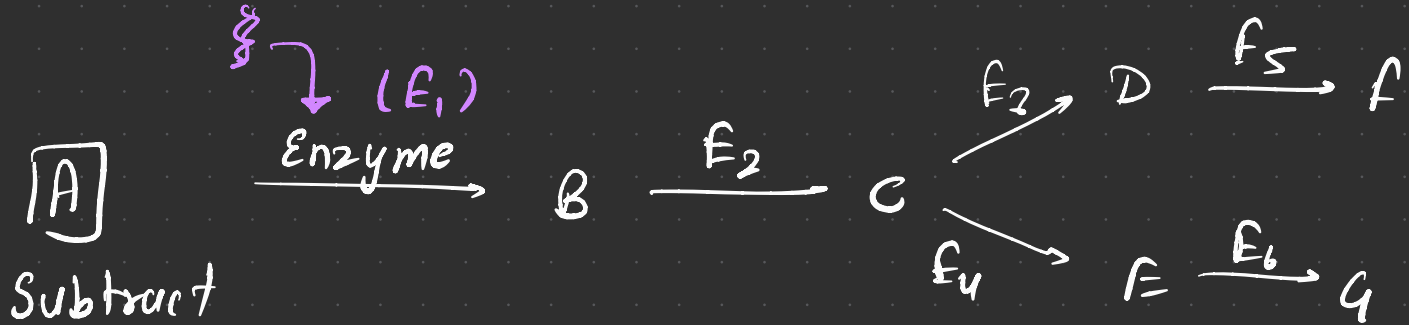
→ ③ no. cannot cross ④ no.

Pleiotropy

→ When 1 factor / gene control more than 1 phenotypic effect



Eg

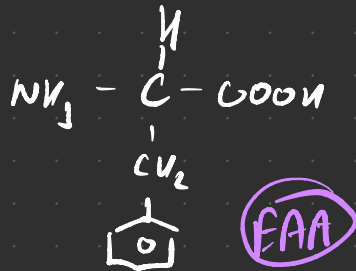


In many Metabolic pathway, gene for initial enzyme synthesis are pleiotropic gene.

Mutation in pleiotropic gene $\rightarrow$ Many effect $\rightarrow$ Many Symptom occur

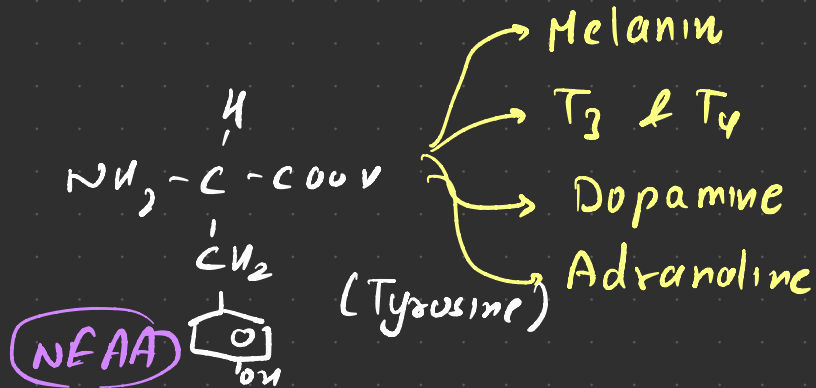
Eg Phenyl ketone Urea / PKU $\rightarrow$ Genetic disorder (Syndrome)

Phenyl alanine



Gene Pleiotropic

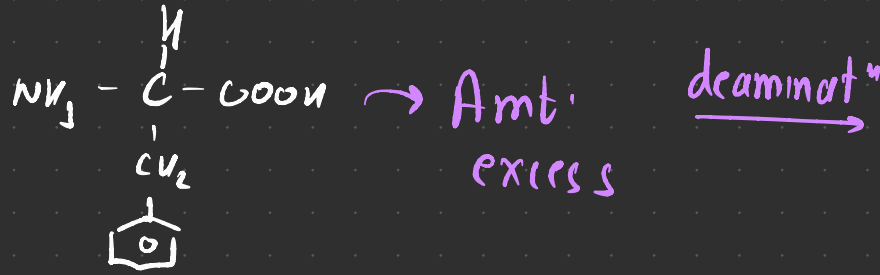
Phenyl alanine
 $\xrightarrow{\text{Hydroxylase}}$



In PKU → Mutation in gene for hydroxylase → Enzyme not

Phenyl alanine

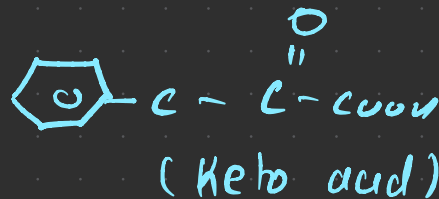
↓
NO
conversion



Phenyl pyruvic acid → Poor excretion in kidney

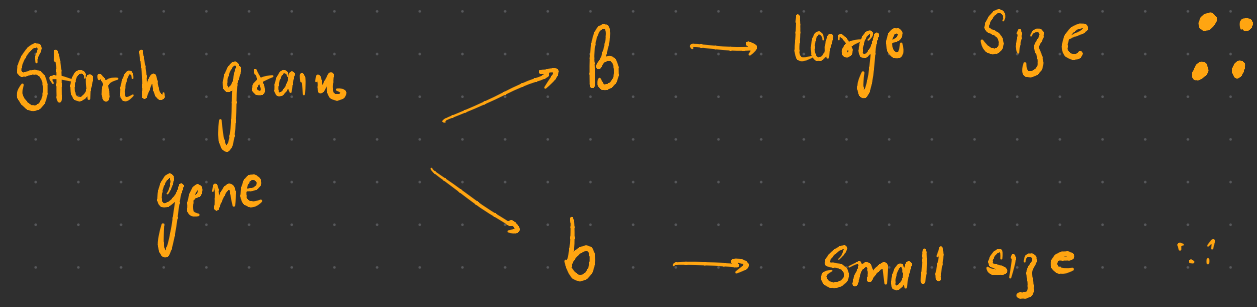
↓
Phenyl Pyruvic acid

↓
Ketone
bodies in
urine



→ Mental Retardation

Starch grain Synthesis gene



→ Starch grain gene → Incomplete dominance

BB → large size
Bb → Medium size
bb → small size

Gene B → 2 trait
(Pleiotropic) Control
↓
Dominance Incomp Dom

Seed Shape = dominance

$BB \rightarrow$ Round

$Bb \rightarrow$ Round

$bb \rightarrow$ wrinkled.

* Dominance is not an autonomous feature of gene

↓

depend upon particular phenotype we choose to examine.

Why some allele are dominant & some are recessive

Gene $\rightarrow$ code for protein $\rightarrow$ trait express

 unmodified allele
(dominant)

Mutation

Modified allele

(option available)

$\rightarrow$  Protein Synthesis

$\rightarrow$ This protein is a functional Enzyme

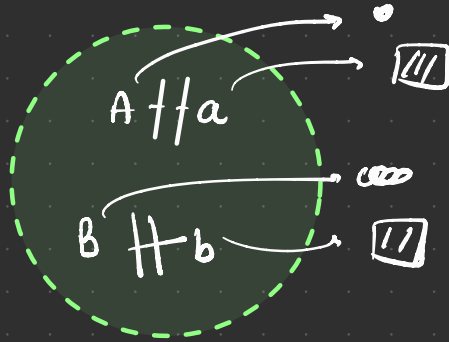
$\downarrow$ substrate
Pdt (Express itself)

 functional Enzyme $\rightarrow$ Trait express] Modified, unmodified

less efficient FE

Not functional (E) $\rightarrow$ Not express itself $\nearrow$ Recessive allele

Gene interaction - 2 gene locus (Dihybrid cross)



Independent assortment - 2 locus
↓
Dihybrid cross involves
follow Mendelism

(PA) $\rightarrow 9:3:3:1$

Polygenic inheritance - (P) Ratio change

Epistasis

Complimentary gene

Supplementary gene

Duplicate

} deviation
of
Mendelism.

Law of independent assortment

Segregation of 1 pair is independent to segregation of another pair

→ Max^m combination forms.



Law of independent
assortment applicable.

{ Both gene locus T & R
on diff chromosome }

↳ 4 type of
Gamete $\begin{matrix} \text{—} & TR \\ \text{—} & tR \\ \text{—} & Tr \\ \text{—} & tr \end{matrix}$



Linkage

↓
no independent
assortment

↓
2 type of gamete

$\textcircled{TR}$

$\textcircled{tr}$

Polygenic inheritance

- 2 or more gene locus Control Single trait
- Each gene locus show contribution
- Skin colour (3 gene locus)

Qualitative inheritance acc. to Mendel

$TT \rightarrow 1m$

$Tt \rightarrow 1m$

$tt \rightarrow 0.50m$

No. of dominant allele $\rightarrow$ not matter

+ve of 1 or 2 dominant allele
 $\downarrow$
same

Quantitative inheritance

$TT \rightarrow 1m$

$Tt \rightarrow 0.75m$

$tt \rightarrow 0.50m$

Matters.

diff. $\textcircled{P}$

① Human Skin colour

Gene $\rightarrow$ Melanin
(A, B, C)

(Mula b.)
 $AaBbCc$

$AA BB CC$ (Darkest skin) Negro

$aa bb cc$ (lightest skin) Albino

(F_1)

② Human IQ

③ Milk production in animal

④ Human height

⑤ wheat kernel colour.

Pleiotropy

1 locus



Many effect

Multiple allele

1 locus

(in more than 1 individual)



MA

Polygenic



Many

locus



1 trait control.

Wheat kernel $\rightarrow$ 2 gene locus $\rightarrow$ 5 (P) Max^m

AA BB x aa bb
Red white

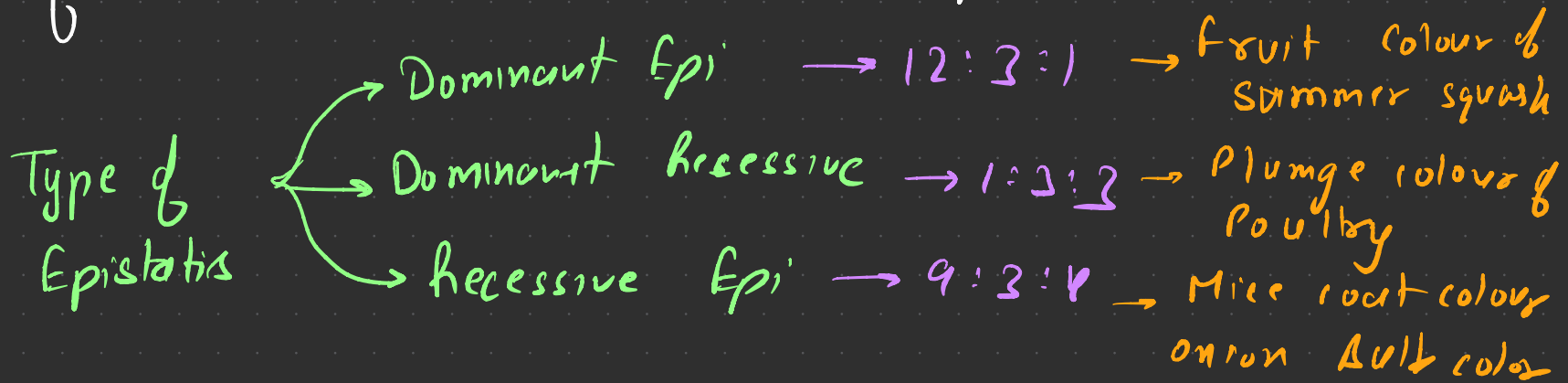
↓
Aa Bb
(intermediate Red)

↓
(P) 1 : (4) : 6 : 4 : 1
 ↓
Aa bb - 2
aa Bb - 2 } (4)

Epistasis

If 'A' gene prod mask/suppress a prod = Dominance.

But if allele (A) of one locus mask/suppress allele of another locus $\leftarrow$ is Epistasis



Complementary gene $\rightarrow 9:7 \rightarrow$ sweet pea
(flower color) $\begin{matrix} \text{Pur} \\ \text{whh} \end{matrix}$

Supplementary gene $\rightarrow 9:3:4 \rightarrow$ Lab Lab (colour of pulses)

Reason why Mendel work not recognized?

① Communication

② Used maths

③ Mendel said $\rightarrow$ Factor discrete unit $\rightarrow$ 1 factor control trait.

④ Physical Basis of inheritance of missing.
(Chromosome not discovered)

Timeline

1831 (Nucleus)

1865 — Mendel work Publish

1880 — Chromosome detail



1900 — Rediscovery of his work



1902 — Chromosome behaviors
in meiosis — study



Sutton & Boveri

Chromosomal Theory

1902 - Sutton & Boveri - Explained Mendel's law/
behaviour of factor
by using behaviour of chromosome
in meiosis.

Segregation of pair of chromosome also segregate
pair of factor.
(Because factor located on
chromosome)

Experimental Verification of CT

↳ TH Morgan (Drosophila)

↳ father of experimental genetics

↳ linkage discovered

↳ Sex "

fruit fly

$2n = 8$

complete life cycle - 2 weeks

large no. of progeny

(Drosophila)

♂ & ♀ easily recognised

features

can easily be grown in artificial medium

easily visible under low power microscope

Test cross

$TtRr \times ttrr$

↓

$TtRr$ - tall Round — Parental (50%)

$Ttrr$ - tall wrinkled

$ttrr$ - dwarf round

$ttrr$ - dwarf wrinkled — Parental

} Recombinant (50%)

Dihybrid cross

Tall Round

dwarf wrinkled

$TT RR$

$\times$

$tt rr$

$\downarrow$

$Tt Rr$

(F_1)

$\downarrow$ selfing

Parental

$\frac{10}{16}$

9

tall Round

3

tall wrinkled

Recombinant

$\frac{6}{16}$

3

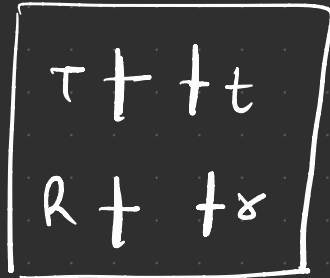
dwarf Round

1

dwarf wrinkled

linkage & Recombination

↓
when 2 factors of diff locus + nt on same chromosome

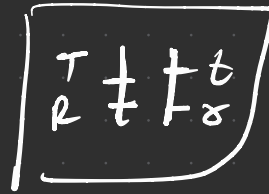


no linkage

↓
Independent assortment

TR
tr

Tr
tR



linkage → Independent assortment
xx

↓
diff. combinatⁿ of factor ↓

TR
tr

Result of linkage.

$\begin{matrix} A+ \\ B+ \end{matrix} \begin{matrix} +a \\ +b \end{matrix}$

no independent
assortment

$\downarrow$
 $\begin{matrix} AB \\ ab \end{matrix}$

$\left\{ \begin{array}{l} \text{Test cross} \rightarrow 1:1:1:1 \\ \text{Normal dihybrid} \rightarrow 9:3:3:1 \end{array} \right.$

$\rightarrow$ Due to linkage $\rightarrow$ not this result.

linkage using test cross

linkage -nt

$\begin{array}{c} TtRr \\ rrrr \end{array} \rightarrow TtRr \times ttrr$

$TtRr$ - tall round

$Ttrr$ - tall wrinkled

$ttrRr$ - dwarf round

$ttrr$ - dwarf wrinkled

Recombinant (50%)

Parental $\rightarrow$ 50%

linkage +nt

$\begin{array}{c} TR \\ tr \end{array} \times \begin{array}{c} Tr \\ tr \end{array}$ (NO crossing over)

(Tall round) $TtRr$

(dwarf wrinkled) $ttrr$

Parental = 100%
 recombinant
 0% Recombinant

Due to linkage Parental type ↑
 Recombinant type ↓

What is crossing over?

$\begin{array}{c} T \\ R \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} t \\ r \end{array}$

G₁ Phase $\xrightarrow{S \text{ phase}}$

Sister Chromatid

$\begin{array}{c} T \\ R \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} T \\ R \end{array}$

$\begin{array}{c} t \\ r \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} t \\ r \end{array}$

G₂ Phase

Crossing Over

$\begin{array}{c} T \\ R \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} T \\ r \end{array}$

$\begin{array}{c} t \\ r \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} T \\ r \end{array}$

$\begin{array}{c} TR \\ Tr \\ tR \\ tr \end{array}$

Meiosis - Prophase I

(Pachytene)

No crossing over

$\begin{array}{c} T \\ R \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} T \\ r \end{array}$

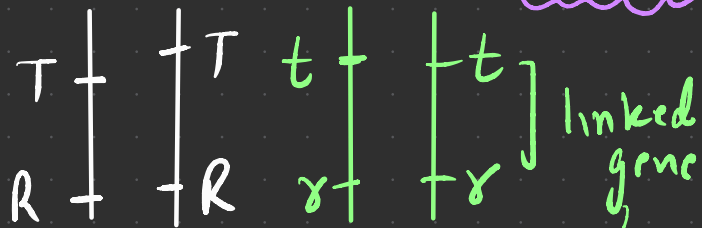
$\begin{array}{c} t \\ r \end{array} \begin{array}{c} + \\ + \end{array} \begin{array}{c} t \\ r \end{array}$

$\begin{array}{c} TR \\ tr \end{array}$

(crossing over)

b/c Non
Sister
chromatid

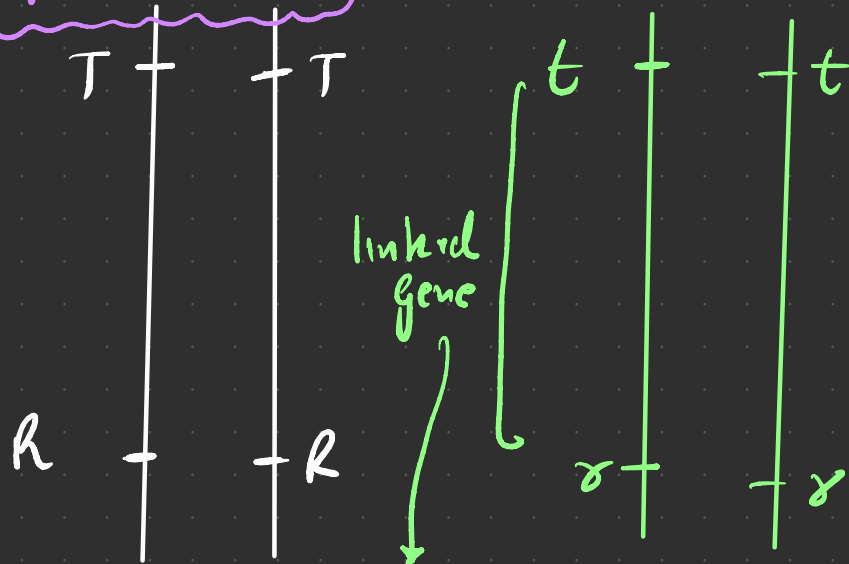
distance $\propto$ Recombinant



less distance

↓
More linkage

less chance of CO



distance More

↓
Linkage less

↓
CO More

Independent
assortment

(P) 50%

(R) 50%

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

distance $\propto \text{Recombinant} \propto \frac{1}{\text{parental}}$

Complete
linkage

(P) 100%

(R) 0%

$\propto \frac{1}{\text{parental Recombinant}}$

incomplete
linkage

P-80%

R-20%

↓

More
distance
b/w linked
gene

P-90%

R-10%

↓

less
distance

linkage Map

Alfred Sturtevant — Student of Morgan — Related
recombinant frequency with relative distance b/w
linked gene

% of recombinant = 1 Map unit = 1 centimorgan

↪ $R = 20\%$
↪ distance = 20

$\begin{matrix} T \\ R \end{matrix}$	$\begin{matrix} t \\ r \end{matrix}$
--------------------------------------	--------------------------------------

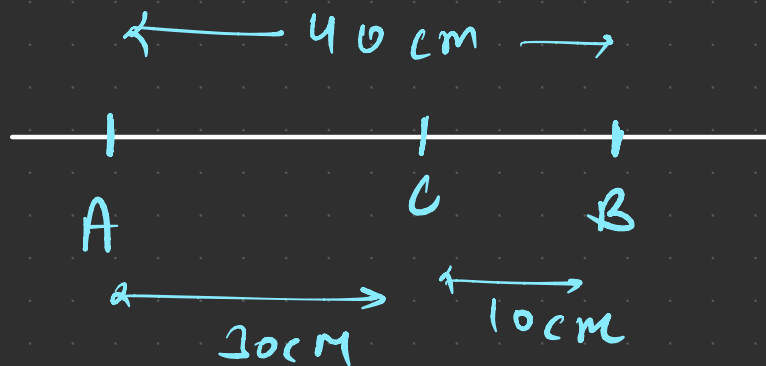
Alfred $\rightarrow$ ① Recombinant freq. $\propto$ Distance b/w linked gene

② He also prepared linkage map where relative position of Gene predict.

Q $A \text{ \& } B = 40'.$

$B \text{ \& } C = 10'.$

$A \text{ \& } C = 30'.$



Sequence = ACB

Q If distance b/w 2 linked gene is 20 cM, then
 give % of Aabb in test cross?

$\begin{array}{c} a \\ b \end{array} \begin{array}{|l} \hline \\ \hline \end{array} \begin{array}{c} A \\ B \end{array} \quad \left. \vphantom{\begin{array}{c} a \\ b \end{array}} \right\} 20 \text{ cM} \rightarrow \underline{20\% \text{ Recombinant}}$

$AaBb \times aabb$

$\downarrow$
 $\begin{array}{c} AaBb \\ \leftarrow \textcircled{Aabb} \\ aAAb \\ aabb \end{array} \quad \begin{array}{c} \text{---} \textcircled{P} \\ \text{---} \textcircled{R} \\ \text{---} \textcircled{P} \end{array}$
 $10\% \rightarrow 20\% = \textcircled{R}$

$TtRr \times ttrr$
 $\downarrow$

$\begin{array}{c} TtRr \\ ttrr \end{array} \quad \left. \vphantom{\begin{array}{c} TtRr \\ ttrr \end{array}} \right\} \textcircled{P}$

$\begin{array}{c} TtRr \\ ttrr \end{array} \quad \left. \vphantom{\begin{array}{c} TtRr \\ ttrr \end{array}} \right\} \textcircled{R}$

Linkage Group

- Gene +nt on 1 Chromosome pair constitute 1 linkage grp
- Gene of 1 linkage grp give recombinant less than 50%.

Human $\rightarrow \sigma \rightarrow 24$ type of chromo
 $\rightarrow \text{♀} \rightarrow 23$ type of chromo

Pea $\rightarrow 2n = 14 \rightarrow \text{grp } (7)$

Maize $\rightarrow 2n = 20 \rightarrow (10) \text{ grp}$

Drosophila $\rightarrow 2n = 8 \rightarrow (4) \text{ grp}$

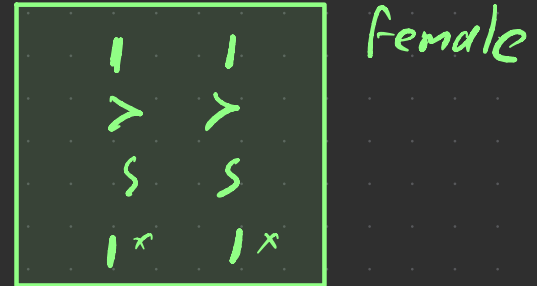
Drosophila Genetics

Linkage — TH Morgan → *Drosophila* (2n=8)
(fruit fly)

Normal dihybrid cross
perform

Expected ① 9 : 3 : 3 : 1

But in reality result deviate this
expected due to linkage.



Morgan perform crosses in which trait + trait on x chromosome

Wild type

$\boxed{D}$

$\boxed{R}$

3 Chr. on x chromosome

Body colour

Eye colour

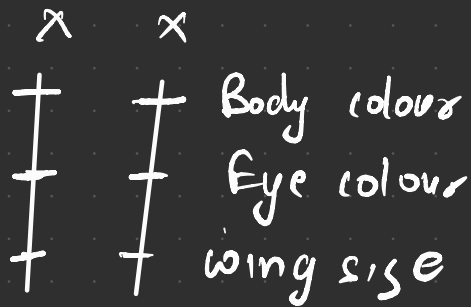
Wing size

Brown (y^+) yellow (y)

Red (w^+) white (w)

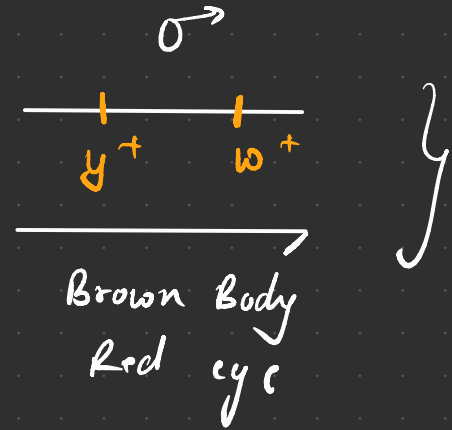
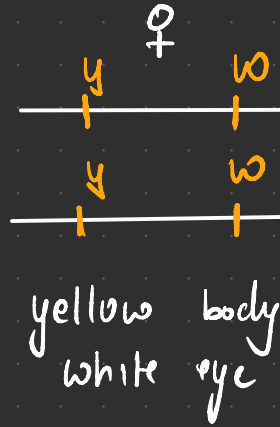
Normal
(m^+)

Miniature
(m)

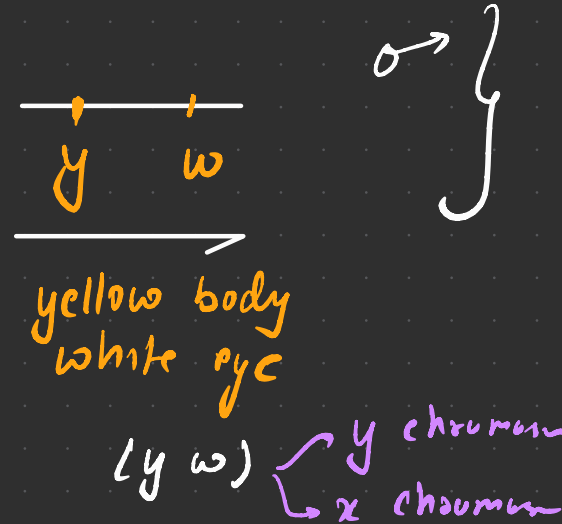
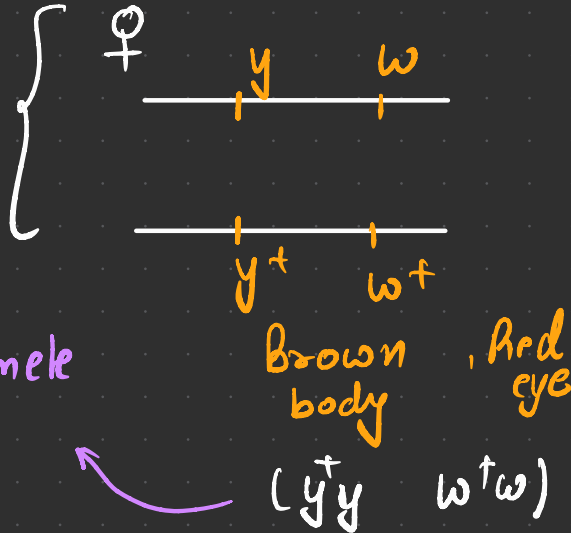


Cross I

Parental



F₁



y⁺w⁺
y⁺w
yw⁺
yw

← y gamete

↓ Intercross

$y^+ w^+$

— ↘

$y w$

$y^+ w^+$

$y w$

$y w^+$

$y w$

$y^+ w$

$y w$

$y^+ w^+$

linkage ↑↑
↑

→ Recombinant

1.3%

$y^+ w$

$y w^+$

$y w$

Cross II

Recombinant

37.2%

Eye Colour

Wing Size



LF.

↓ Inter cross





⇓
cross I

⇓
 $R = 1.3\%$

⇓
cross II

⇓
 $R = 37.2\%$

Mutation

Any sudden inheritable change in DNA seq.

Source of variation

by devices

Plant → Evening Primrose

2 type

— Somatic



Skin cell

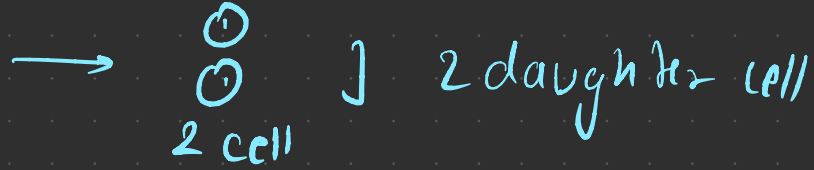
Liver cell

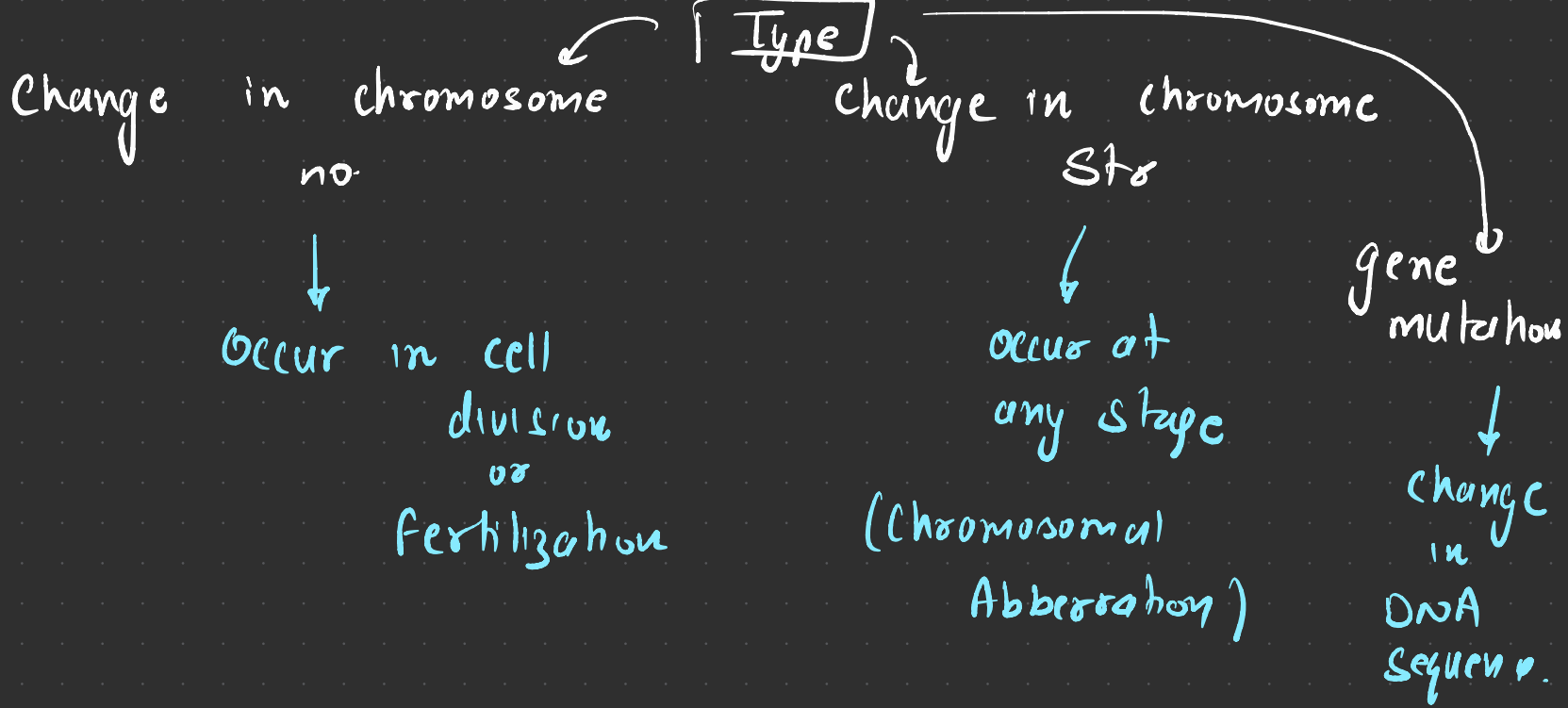
— Gametic



Mutation in germ cell ⇒

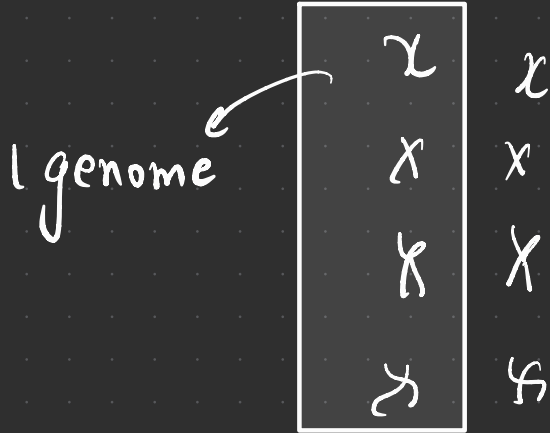
not goes to next gen





Genome

Haploid set of chromosome



In human
" 2 3 chromosome
genome

Change in chromosome no.

Poly ploidy

More than 2 copy of genome

Naturally → Tetraploid Endosperm
→ Hexaploid wheat

Aneuploidy

one or two chromosome

↑ or ↓

human (47) → $2n+1$

Trisomy

Tetrasomy (48) $2n+2$

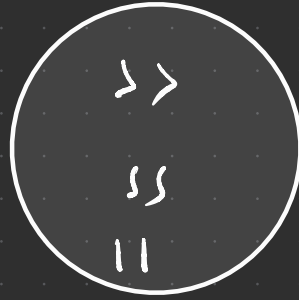
Patau's → 13th

Down's → 21st

Edward → 18th

Monosomy (46) $2n-1$

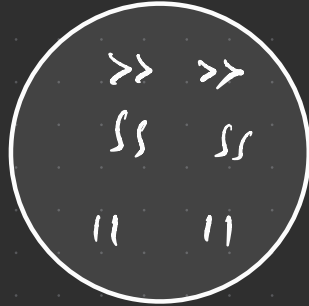
Nullisomy $2n-2$ (45)



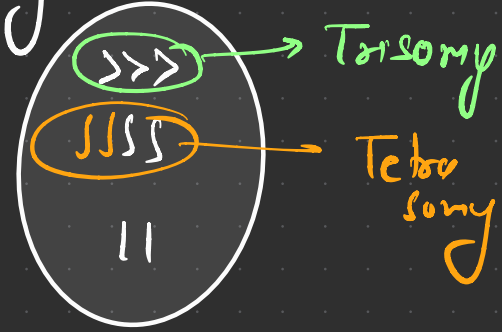
Normal



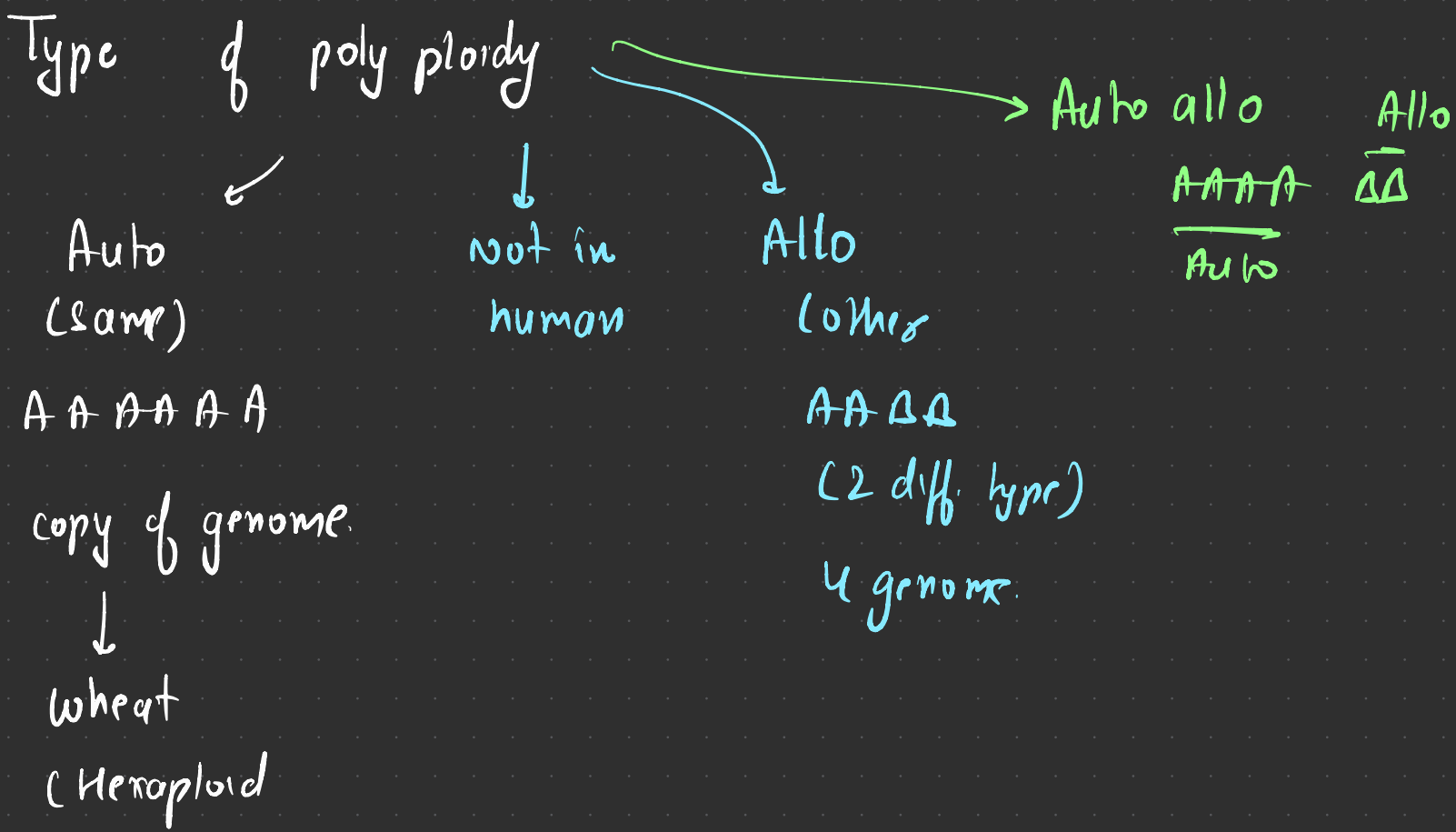
Polyploidy



Aneuploidy



(2 copy of genome)



Change in Chromosome Structure $\Rightarrow$ due to Mutagen
cause mutation



deficiency



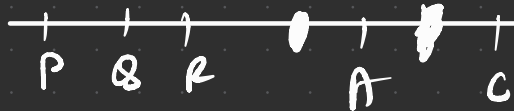
inversion



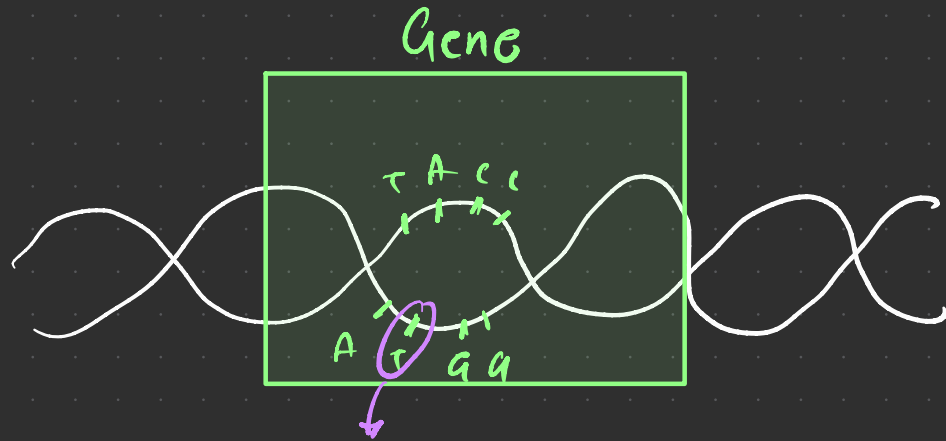
duplication



deletion



Gene mutation



change in single nucleotide

(Point mutation)

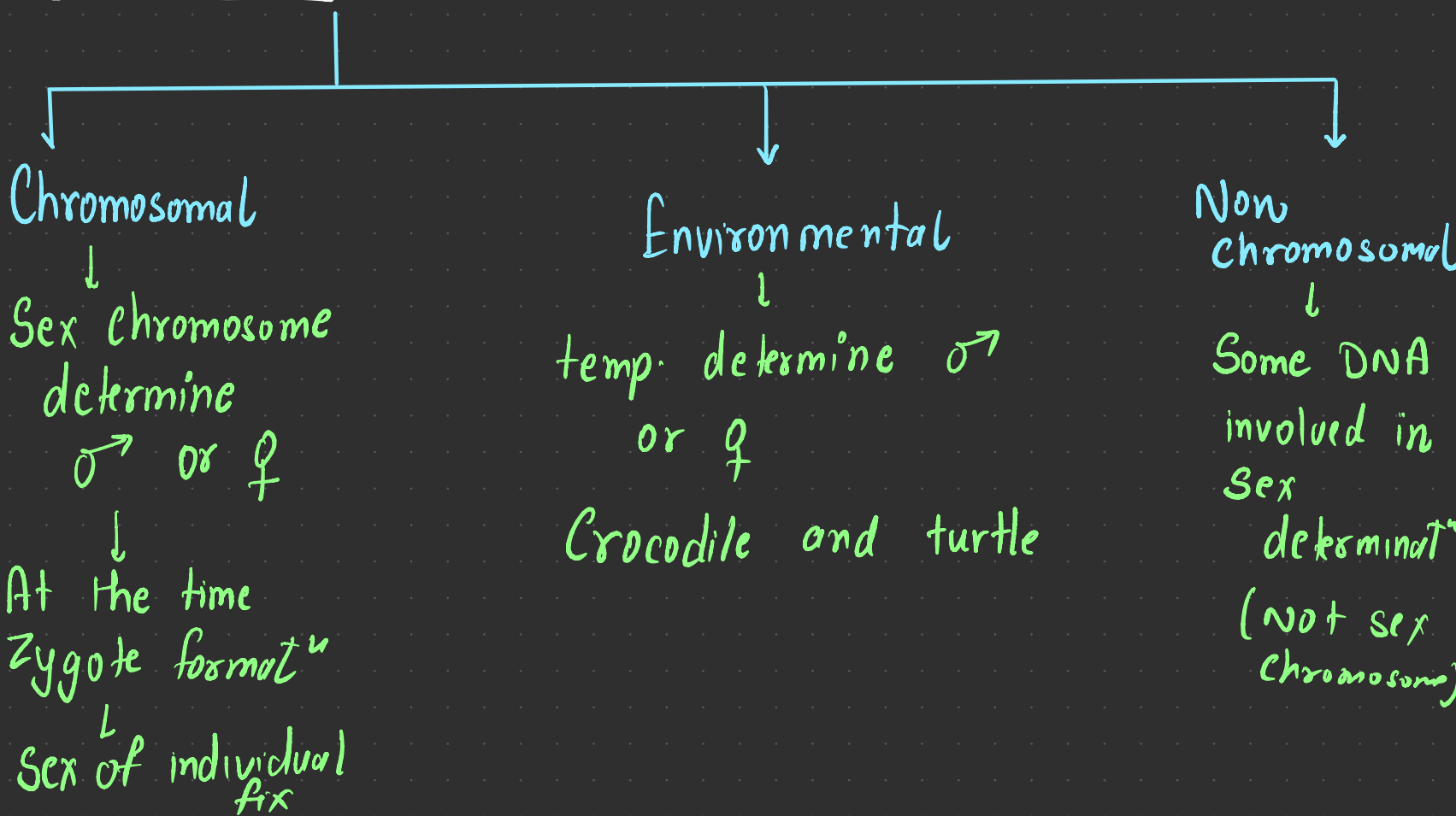
Addⁿ

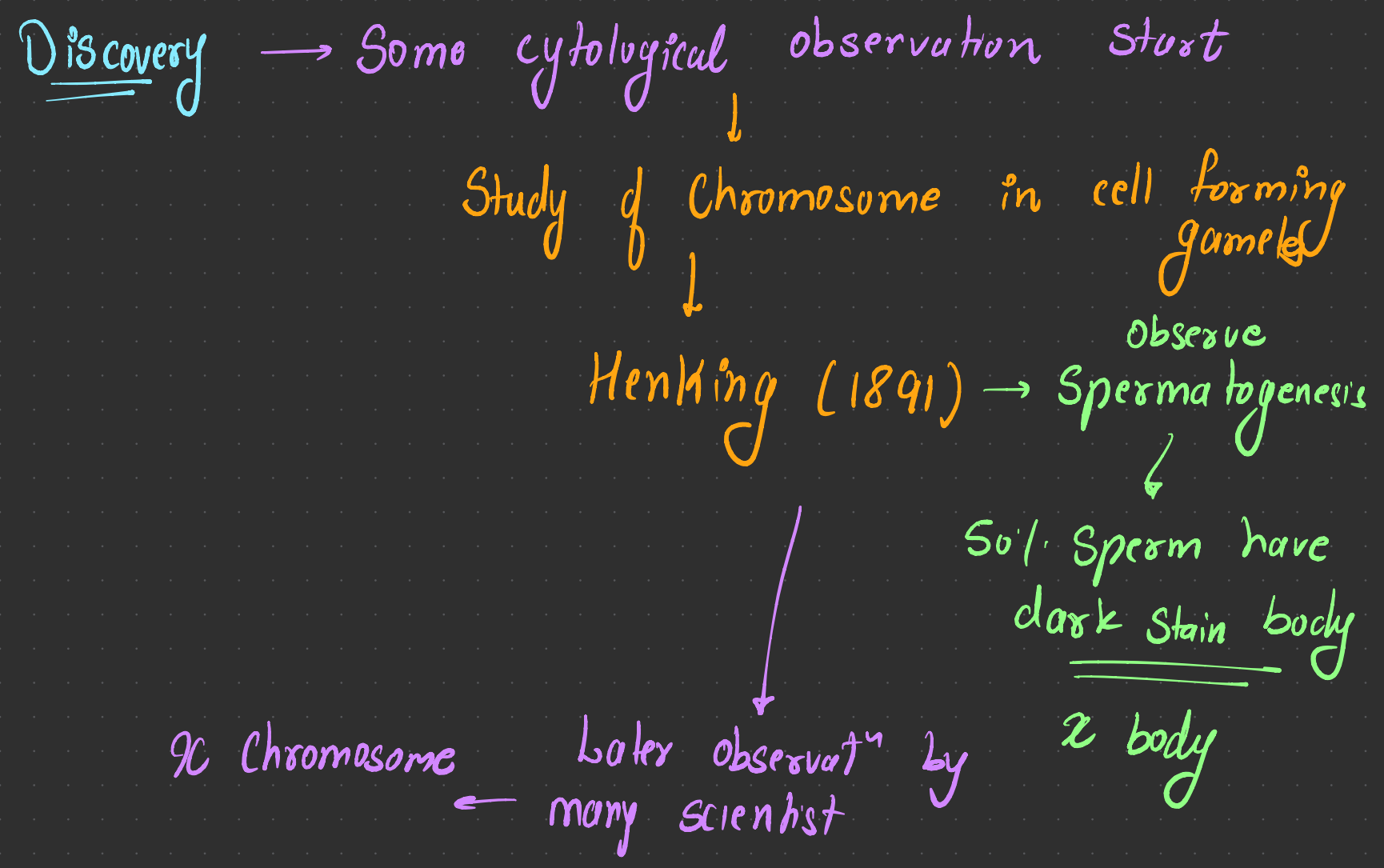
deletⁿ

Substitutⁿ

Sickle cell anaemia

Sex determination

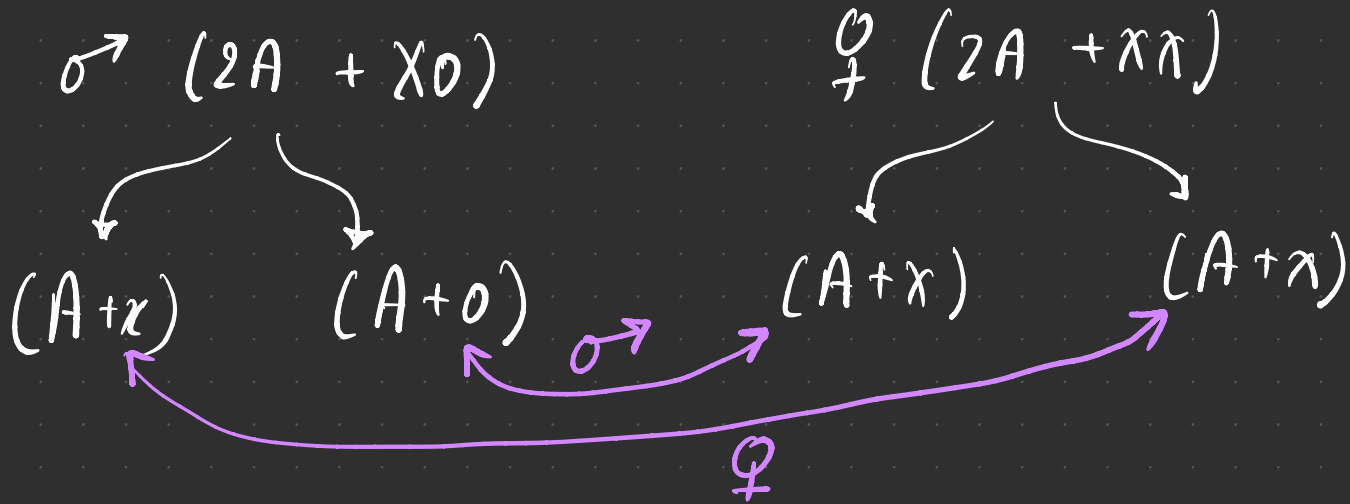




Later it was observed, the role of x Chromosome in Sex determinatⁿ



x Chromosome $\Rightarrow$ Sex Chromosome



$XX - XO$ type (XO type) $\rightarrow$ 1st time discovered $\rightarrow$ Insects
(Grasshopper)

σ^7 (Heterogamete)
 $(2A + XO)$
 $\wedge$
 $(A+X) \quad (A+O)$

ϕ
 $(2A + XX)$
 $\wedge$
 $(A+X) \quad (A+X)$

$xX - xY$ type $\rightarrow$ Drosophila & Human

♂

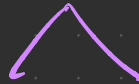
$(2A + XY)$



$(A + x)$ $(A + y)$

♀

$(2A + XX)$



$(A + x)$ $(A + x)$

ZZ - ZW  Birds

♂

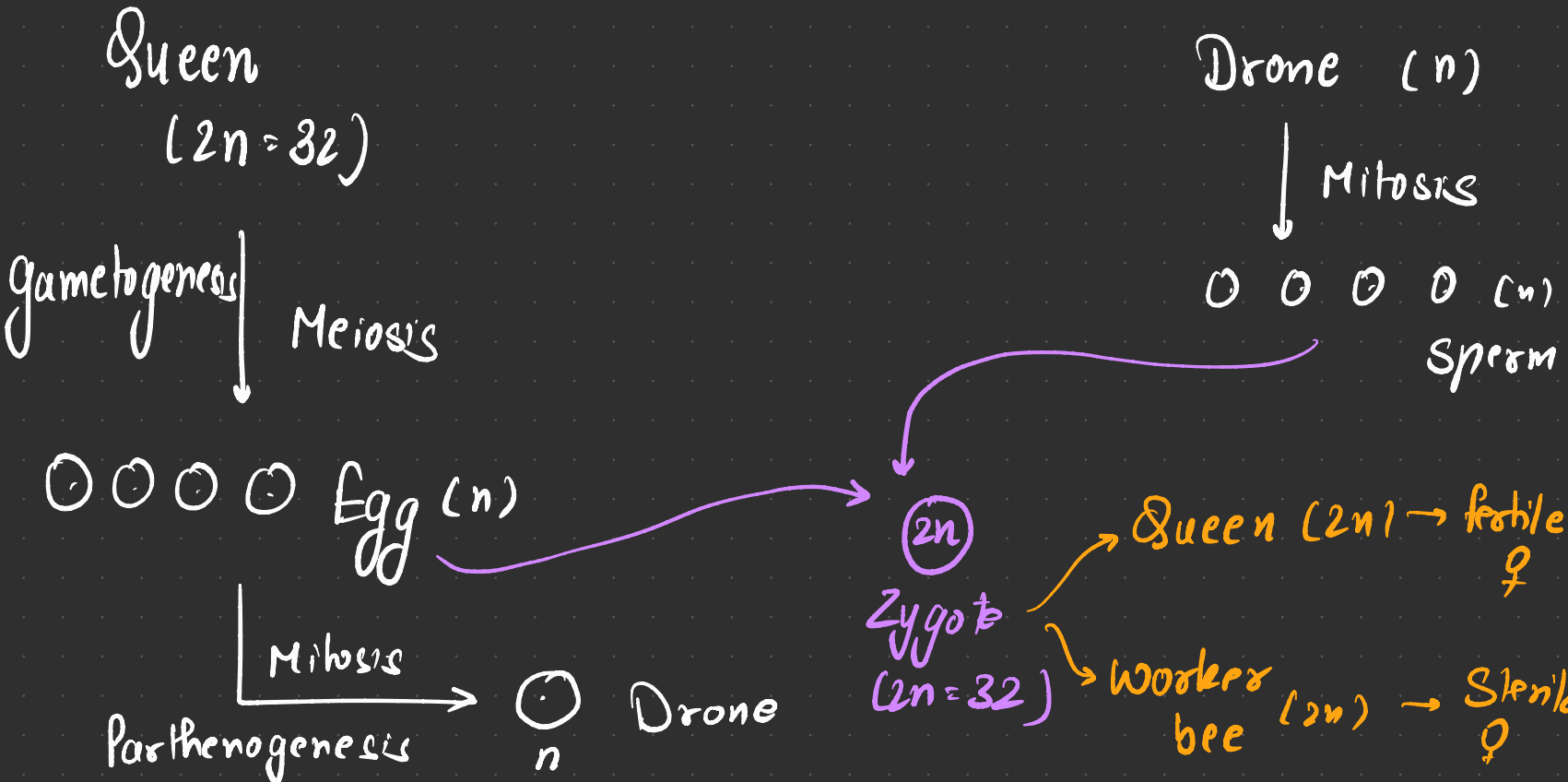
↓

♀

Heterogametes

Haplodiploidy

Social insects $\begin{cases} \text{Queen} \\ \text{Worker} \\ \text{Drone} \end{cases}$



Pedigree analysis

Autosomal
Trait

^
Dominant Recessive

Sex trait

↓
trt on x
chromosome

♀ → xx

♂ → x

Eg Haemophilia
Colour Blindness

Y linked

↓
only in
♂

(Holoandric)

↓

Hypertrichose

(Hair on
Pinna)

TDF
(Teste determining^{ter})

Sex limited

♀

♂

gene +nt on both autosome

only express in one sex

→ Breast development

Sex influenced

Pattern baldness

↓

+nt on autosome

↓

Express in both sex, more in one

♂ → Baldness + thinning of Hair

♀ → thinning of Hair

Meaning → Study of Genetic trait (Disease or Non disease)

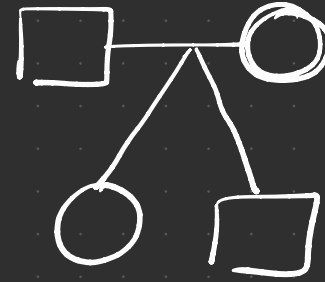
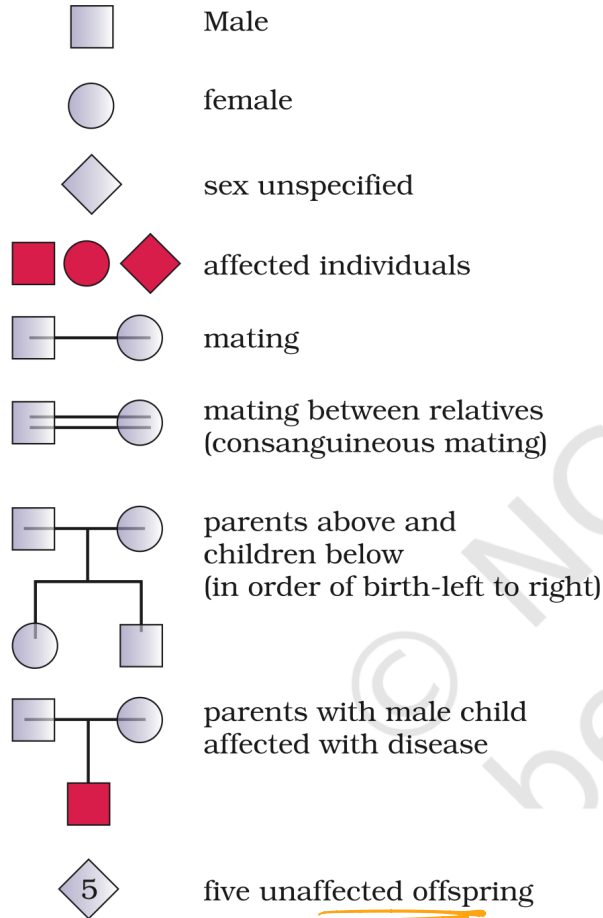
Req → In human it is not possible to perform control cross or back cross.

Study of any trait → family history

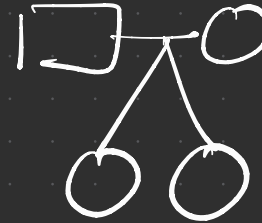
use

- ① Study genetic disease
- ② Predict genetic disorder

Symbol



Dizygotic
twin



Monozygotic
twin

 } carriers $\rightarrow XX^h \rightarrow$ ♀ normal but carry
 gene for haemophilia

 ♂ carrier

Autosomal Dominant

Autosomal
Recessive

X linked

Y linked

Myotonic
dystrophy

Sickle cell
anaemia

Colour
blindness

Hyper
trichosis

PKU

Hemophilia

Aa
 AA } $\rightarrow$ Diseased
 $aa \rightarrow$ Normal

Thalassemia
Muscular dystrophy
Cystic Fibrosis

Carrier $\rightarrow$ $\left. \begin{array}{c} XX \\ \hline Xx \end{array} \right\}$ Normal
 $x^u x^u$ - diseased

$aa \rightarrow$ Diseased
Carrier $\rightarrow$ $\left. \begin{array}{c} Aa \\ \hline AA \end{array} \right\}$ Normal

Genotypes for trait

Pedigree $\rightarrow$ 2 component

- Symbol $\rightarrow$  
- Genotype $\rightarrow$ AA Aa aa
 XX XY

for trait $\rightarrow$ Autosomal - Genotype

AA Aa aa $\rightarrow$ $\textcircled{A}$ domi
 $\downarrow$
Diseased
 $\boxed{aa}$ $\rightarrow$ Normal

AA Aa aa $\rightarrow$ Normal
 $aa \rightarrow$ Diseased $\rightarrow \textcircled{A}$ depressive.

Sex linked

$\rightarrow \begin{matrix} X \\ Y \end{matrix}$

} genotype

$\rightarrow \begin{matrix} XX \\ XY \end{matrix}$

$X^c X^c \rightarrow$ colour blind
♀

$X^c X \rightarrow$ Normal
(carrier)

$XX \rightarrow$ Normal

$X^c Y \rightarrow$ Diseased
♂

$XY \rightarrow$ Normal

Sickle cell anaemia

(A) Recessive

⊗ → aa

□ Aa, AA

Myotonic dystrophy

(A) D

▤ → Aa, AA

□ → aa

○ → aa

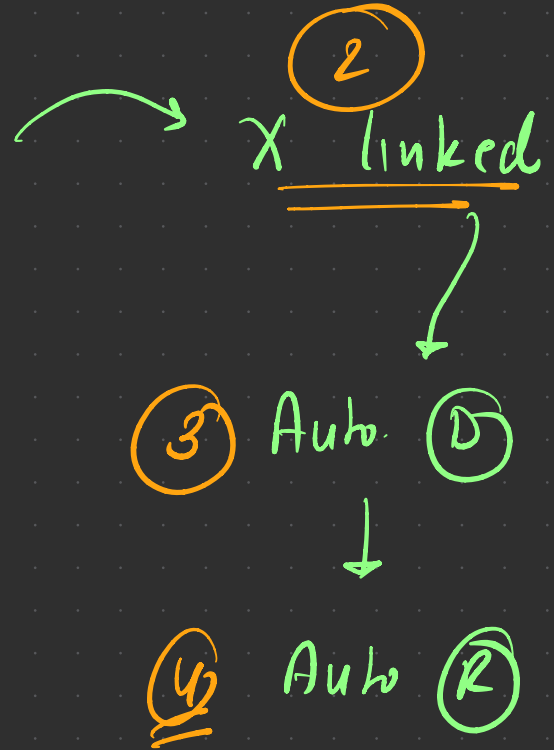
⊙ → Aa, AA

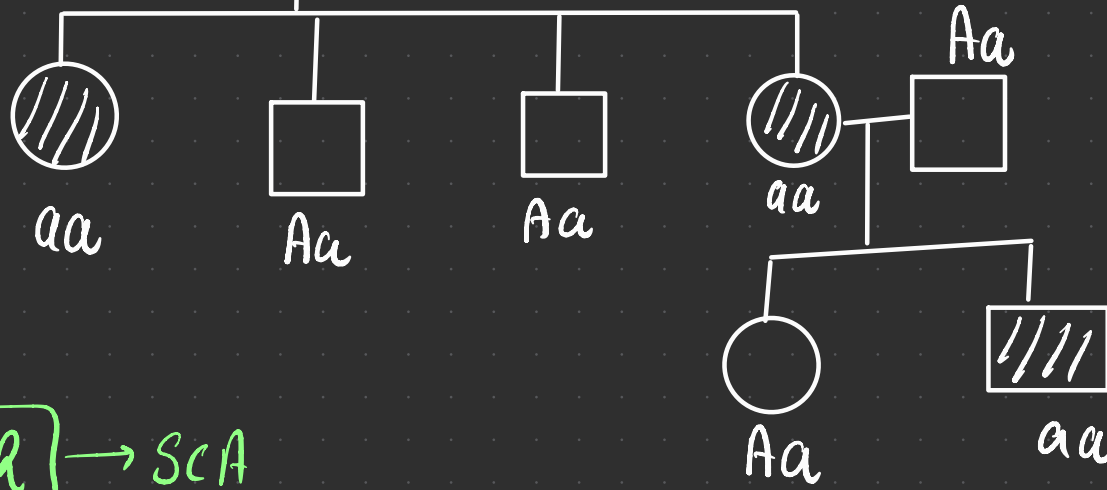
How to check Pedigree

if y linked

①

→ only ♂
affected





→ IA · R → ScA

① A · D x x

② X linked x x

③ Y linked x x

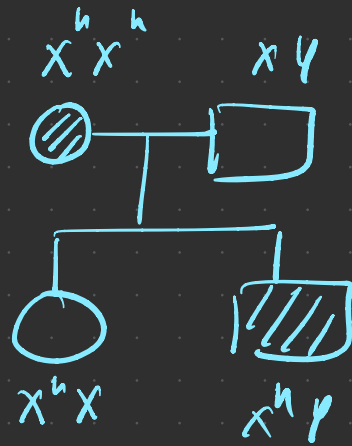
A. Recessive $\rightarrow aa$  

A. Dominant $\rightarrow aa$ unaffected

Sex linked $\rightarrow$ Male always diseased

If ♀ affected $\rightarrow$ all son affected

to affect ♀ $\rightarrow$ father should be affected.



Y-linked $\rightarrow$  only ♂ affected

① Parents normal $\rightarrow$ Progeny affected $\rightarrow$ New $\boxed{A-D}$

② If both affected $\rightarrow$ progeny normal $\rightarrow$ New $\textcircled{A} \textcircled{F}$

③ Y linked $\rightarrow$ only σ affected

Genetic disorder

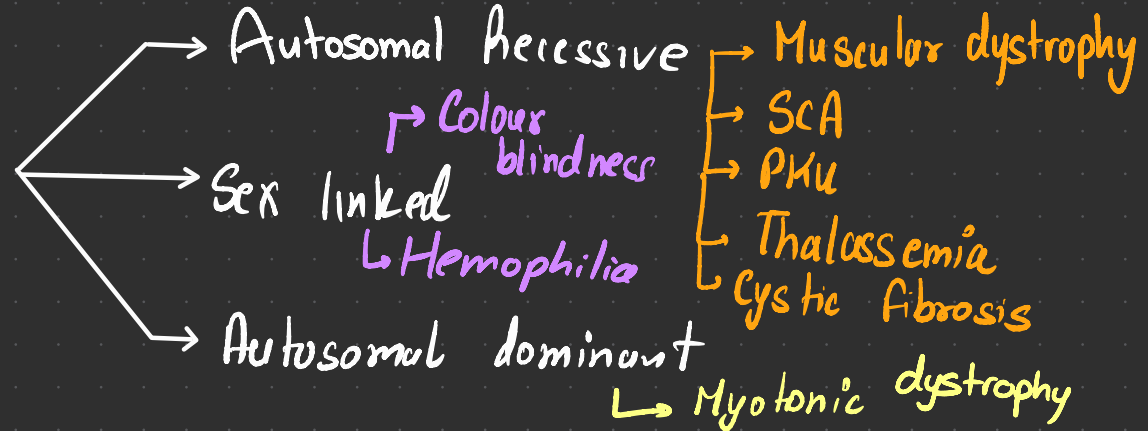
Mendelian disorder

- follow Mendel's law of inheritance
- Single gene mutation
- Pedigree possible

Sickle cell anaemia

Thalassemia

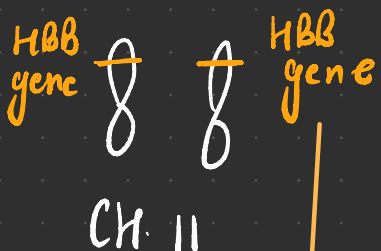
Colour blindness



Chromosomal disorders

- Change in ch. no. or str
- cannot drawn in Pedigree

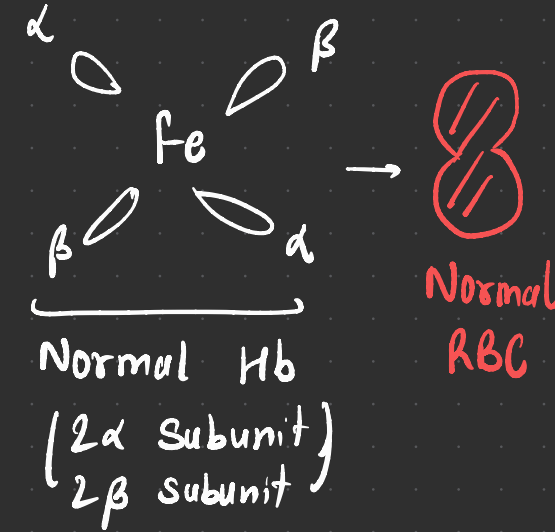
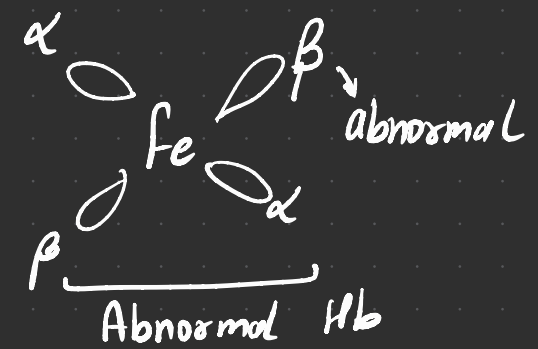
SCA



Protein : (β subunit)
normal

Mutation

Abnormal β subunit



$Hb^A Hb^A \rightarrow$ normal RBC (Biconcave)

$Hb^A Hb^S \rightarrow$ (70% Hb^A) Normal RBC with 30% $Hb^S \xrightarrow{O_2 \downarrow \downarrow}$ Sick shape

↓
Advantage

$Hb^S Hb^S \rightarrow$ Abnormal Hb
↓
death in infant stage

↓
Malaria
advantage

Problem

- Sickle Shape RBC → Block Capillaries
- Blood supply ↓

Symptom

- Severe pain
- Stroke
- Sickle RBC → 20 day

Normal RBC	Point Mutation →	Sickle cell
GAG		GUG
Glutamic acid		Valine

Thalassemia

Less amount of Haemoglobin in RBC



CH-11

→ β subunit



CH-16

→ α subunit



Normal RBC

β thala



Point mutation
→
in both HBB gene

Less amt. of β chain
Hb synthesis -nt



Mutation
→
in 3 allele

Less amt. of α -chain

→ Less amt. of Hb

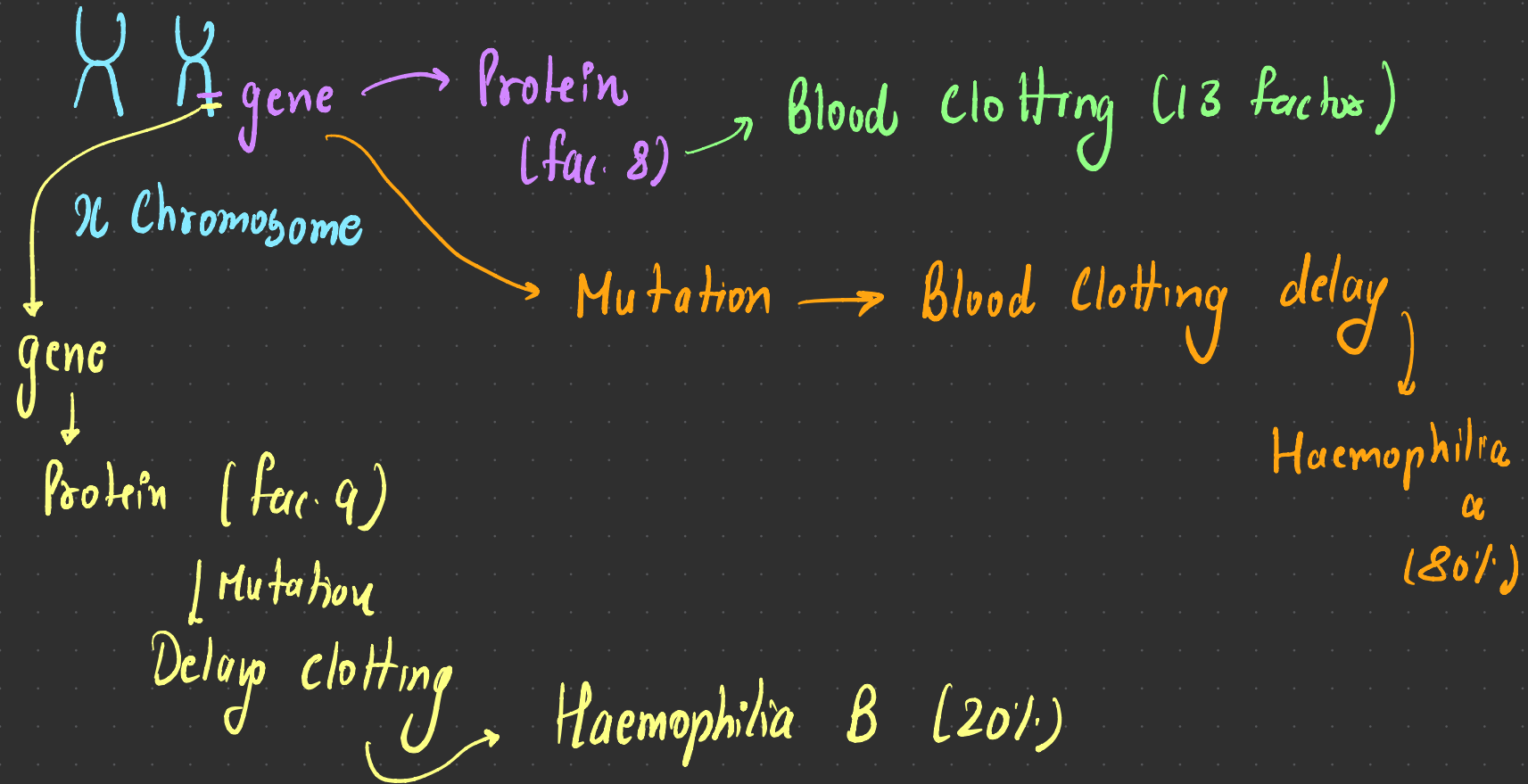
α -thalassemia

less amt. of O_2

Nausea
Fatigue



Haemophilia



$XX \rightarrow \text{normal } \text{♀}$

$XY \rightarrow \text{normal } \text{♂}$

$XX^h \rightarrow \text{carrier } \text{♀} \rightarrow \text{normal}$

$X^hY \rightarrow \text{diseased } \text{♂}$

$X^hX^h \rightarrow \text{diseased}$

Royal Family of England

↓
Queen Victoria

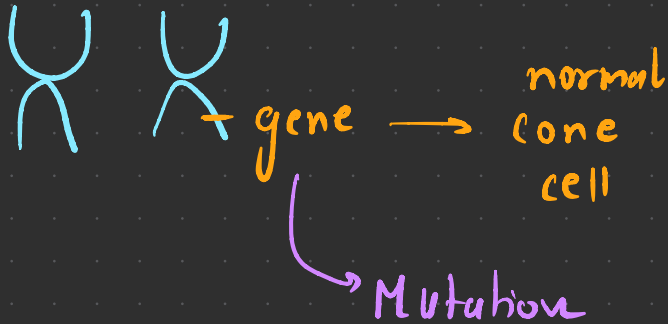
Colour Blindness

→ check by Ishihara test

on Retina → cone + nt

- Red
- Blue
- Green

♂ → 8%
♀ → 0.4%



XX → normal ♀

XX^c → carrier ♀ → normal

X^cX^c → diseased

Chromosomal disorder

Change in number of chromosome $\rightarrow$ Aneuploidy (Trisomy) (due to non disjunction)

Trisomy $\rightarrow$ Patau
 $\rightarrow$ Edward
 $\rightarrow$ Down Syndrome = 47 $\rightarrow$ 21st extra

Klinefelter's syndrome = 47 $\rightarrow$ 44 + XX + Y (not trisomy)

Turner's syndrome = 45 (44 + XO)

Super male $\rightarrow$ (44 + XY + Y)

Super female $\rightarrow$ (44 + XXX)

Down Syndrome

1866 - longdon Down

47 chromosome (21st chromosome)

Symptoms

→ Short Stature

Round face

Mental Retarded

Congenital Heart disease

→ open mouth

→ furrowed
tongue

→ Distinct
palm
crease

→ loop in
fingers

Turner's Syndrome

♀ $(44 + x0) = 45$

→ Short Stature

→ one x less

↓
ovary Rudimentary

↓
no gamete formatⁿ

→ Sterile

Klinefelter's Syndrome

♂ $(44 + xxy) = 47$

→ Gynecomastia

→ Testis underdeveloped

→ Sterile

→ Not Trisomy

→ 10:15 to 11:15
M W F → 11th
T T S → 12th

833 9827528 (1:1)