

PRINCIPLES OF INHERITANCE AND VARIATION

Gregor Mendel experimented on Garden pea or Pisum sativum.

In garden pea Mendel selected seven contrasting characters.

	characters	Dominant	Recessive
1.	Seed colour	Yellow	Green
2.	Seed shape	Round	Wrinkled
3.	Pod colour	Green	Yellow
4.	Pod shape	inflated	constricted
5.	Flower colour	Violet	White
6.	Flower position	Axial	Terminal
7.	Plant height	Tall	Dwarf

→ Mendel selected Pisum sativum because of the following reasons.

1. It is easily available
2. It has a short lifespan
3. Different varieties are present
4. It can be self pollinated

→ Monohybrid Cross

It is the cross involving a single character. Mendel

selected seed colour for

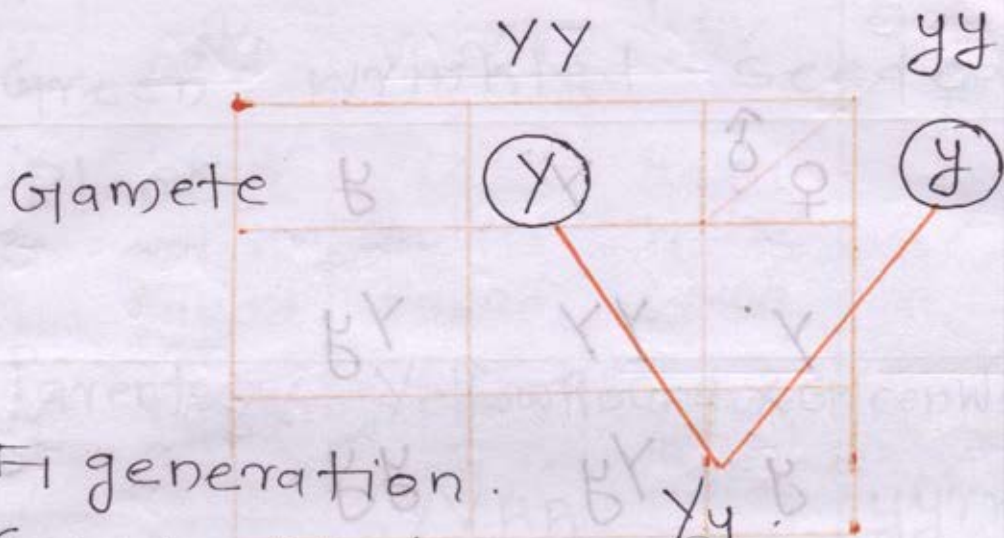
his monohybrid cross. He

crossed a pure breeding

Yellow seed coloured plant

with a pure breeding green seed coloured plant.

Parents. Yellow x Green.



F₁ generation.

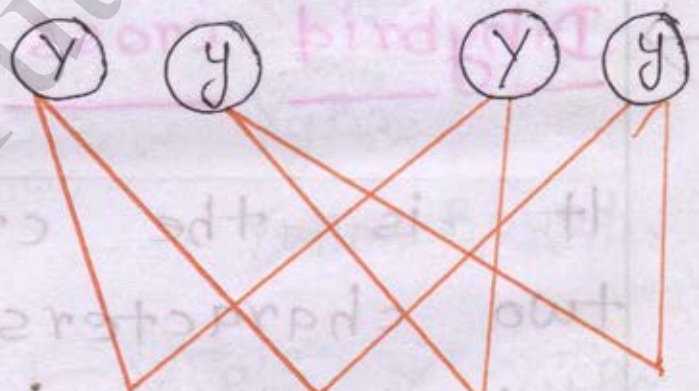
(First filial generation)

Yellow.

F₁ Parent

Yy x Yy

Gamete



F₂ generation

YY Yy Yy yy

Yellow Yellow Yellow Green

→ Punnet square

♀ ♂	Y	y
Y	YY	Yy
y	Yy	yy

Phenotypic ratio: — 3:1

Genotypic ratio: — 1:2:1

→ Dihybrid cross

It is the cross involving two characters. Mendel selected seed colour and seed shape for his dihybrid cross. He crossed a pure breeding yellow round seeded

Plant with a pure breeding
Green wrinkled seeded

Plant

Parents: Yellow Round \times Green Wrinkled

$YYRR$

$yyrr$

Gametes

YR

yr

$YyRr$

F₁ generation.

Yellow Round.

F₁ Parent $YyRr \times YyRr$

Gametes

YR

Yr

yR

yr

YR

Yr

yR

yr

♂ ♀	YR	Yr	yR	yr
YR	YYRR Yellow Round	YYRr Yellow Round	YyRR Yellow Round	YyRr Yellow Round
Yr	YYRr Yellow Round	YYrr Yellow Wrinkled	YyRr Yellow Round	Yyrr Yellow Wrinkled
yR	YyRR Yellow Round	YyRr Yellow Round	yyRR Green Round	yyRr Green Round
yr	YyRr Yellow Round	Yyrr Yellow Wrinkled	YyRr Green Round	yyrr Green Wrinkled

phenotypic ratio :— 9 : 3 : 3 : 1

Genotypic ratio :— 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

YYRR — 1	YyRR — 2	yyRR — 1
YYRr — 2	YyRr — 4	yyRr — 2
YYrr — 1	Yyrr — 2	yyrr — 1

→ Law of Mendels

1. Law of Dominance →

When two factors are involved in a cross one factor will mask the effect of other.

The expressed factor is known as Dominant and other one is Recessive.

2. Law of Segregation.

When two factors are involved in a cross the factor pairs Segregates during the formation of gametes. It is based on monohybrid cross.

3. Law of Independent Assortment

It is based on dihybrid cross. When two characters are involved in a cross the factor pairs —

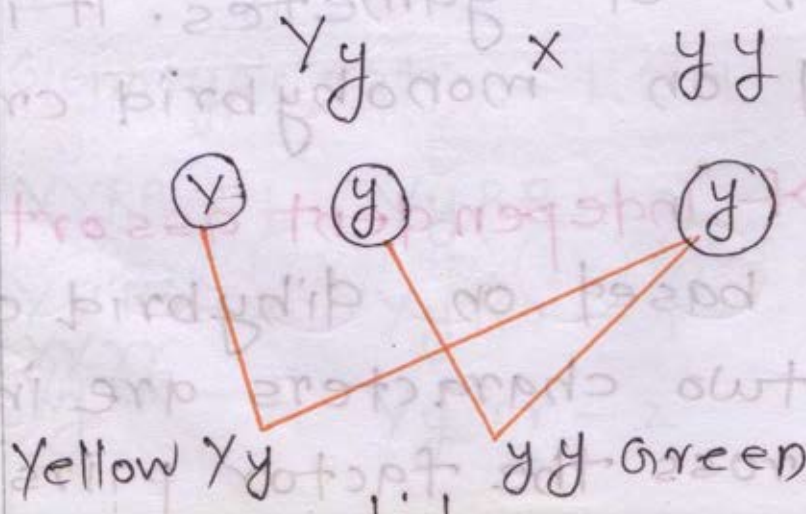
assort independent of each other

→ **Backcross** and **Test cross**.

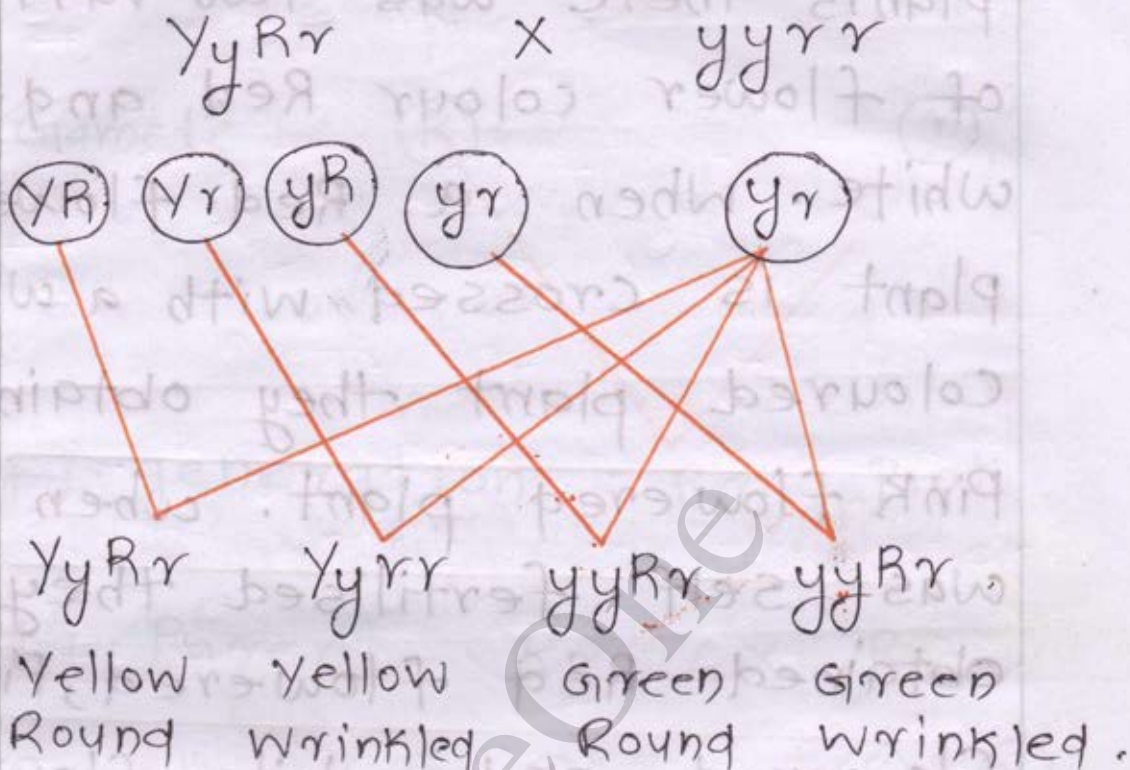
When the F_1 hybrid is crossed with anyone of its parent. It is backcross.

When the F_1 is crossed with its Recessive parent. It is Test cross. The test cross is used to find out unknown genotype of individual.

Monohybrid test cross:



Dihybrid test cross.



→ Other Patterns of Inheritance.
(Non-Mendelian inheritance).

1. Incomplete dominance.

It was observed by Carl Correns in Mirabilis jalapa (4'clock plant) and also in Antirrhinum / snap

dragon / dog flower. In these plants there were two varieties of flower colour Red and white. When a Red flowered plant is crossed with a white coloured plant they obtained Pink flowered plant. When it was self fertilised they obtained Red flowered, Pink flowered and white flowered plant in the ratio of 1:2:1. It is due to Incomplete dominance. The red is incompletely dominant over white.

Parent Red \times White

RR

rr

Gamete

(R)

(r)

F₁ generation Rr Pink

F₁ Parent $Rr \times Rr$

gamete

(R)

(r)

(R)

(r)

RR

Rr

Rr

rr

Red

Pink

Pink

White

1 : 2 : 1

2. Multiple Allelism.

Alleles are copies of genes. When a character is controlled by more than two alleles, it is known as multiple allelism.

Group	Antigen	Antibody	Genes
A	A	a	$I^A I^A, I^A i$
B	B	b	$I^B I^B, I^B i$
AB	A & B	—	$I^A I^B$
O	—	a & b	ii

Blood group is controlled by the alleles I^A, I^B , and i . So it is an example for multiple allelism.

In AB group I^A and I^B express together. So known as Codominance.

→ Chromosomal basis of inheritance

(Sutton and Boveri hypothesis)

Sutton and Boveri observed the behaviour of genes and chromosomes. And observed the following similarities.

1. The genes and chromosomes are found in paired condition.
2. Genes and chromosomes separate during the formation of gamete.

3. The paired condition is restored after fertilization.

4. Genes and chromosomes show independent assortment.

Based on these similarities Sutton and Boveri proposed that chromosomes are the basis of inheritance.

Q A pea plant with green pod coloured and round seed was crossed with yellow pod coloured wrinkled seeded plant. find out the ratios in F_2 .

Parent : Green Round \times Yellow wrinkled

$GGRR$

$ggrr$

Gamete :

(GR)

(gr)

F_1 generation.

$GgRr$

Green Round.

F_1 Parent

$GgRr$

$\times GgRr$

gametes

(GR)

(Gr)

(gR)

(gr)

(GR)

(Gr)

(gR)

(gr)

♀ \ ♂	GR	G _r r	gR	gr
GR	GGRR Green Round	GGRr Green Round	GgRR Green Round	GgRr Green Round
G _r r	GGRr Green Round	GGrr Green Wrinkled	GgRr Green Round	Ggrr Green Wrinkled
gR	GgRR Green Round	GgRr Green Round	ggRR Yellow Round	ggRr Yellow Round
gr	GgRr Green Round	Ggrr Green Wrinkled	ggRr Yellow Round	ggrr Yellow Wrinkled

phenotypic ratio: — 9:3:3:1

Genotypic ratio: — 1:2:1:2:4:2:1:2:1

Linkage

Genes on a single chromosome are said to be linked together. The genes on a single chromosome are inherited into a single gamete. As the distance b/w the genes increases linkage decreases. It is known as incomplete linkage. As the distance b/w genes decreases linkage increases. It is known as complete linkage.

→ Crossing over and Recombination

Crossing over is the overlapping of chromosomal segments. The region where chromosomal

Segments overlaps pair's +

chiasmata - Chromosomal

Segments breaks at

chiasmata exchange of

chromosomal segments

occur when it rejoins.

It is known as recombination.

Recombination increases

Variation. As the distance

blw genes increases linka-

ge decreases and recombi-

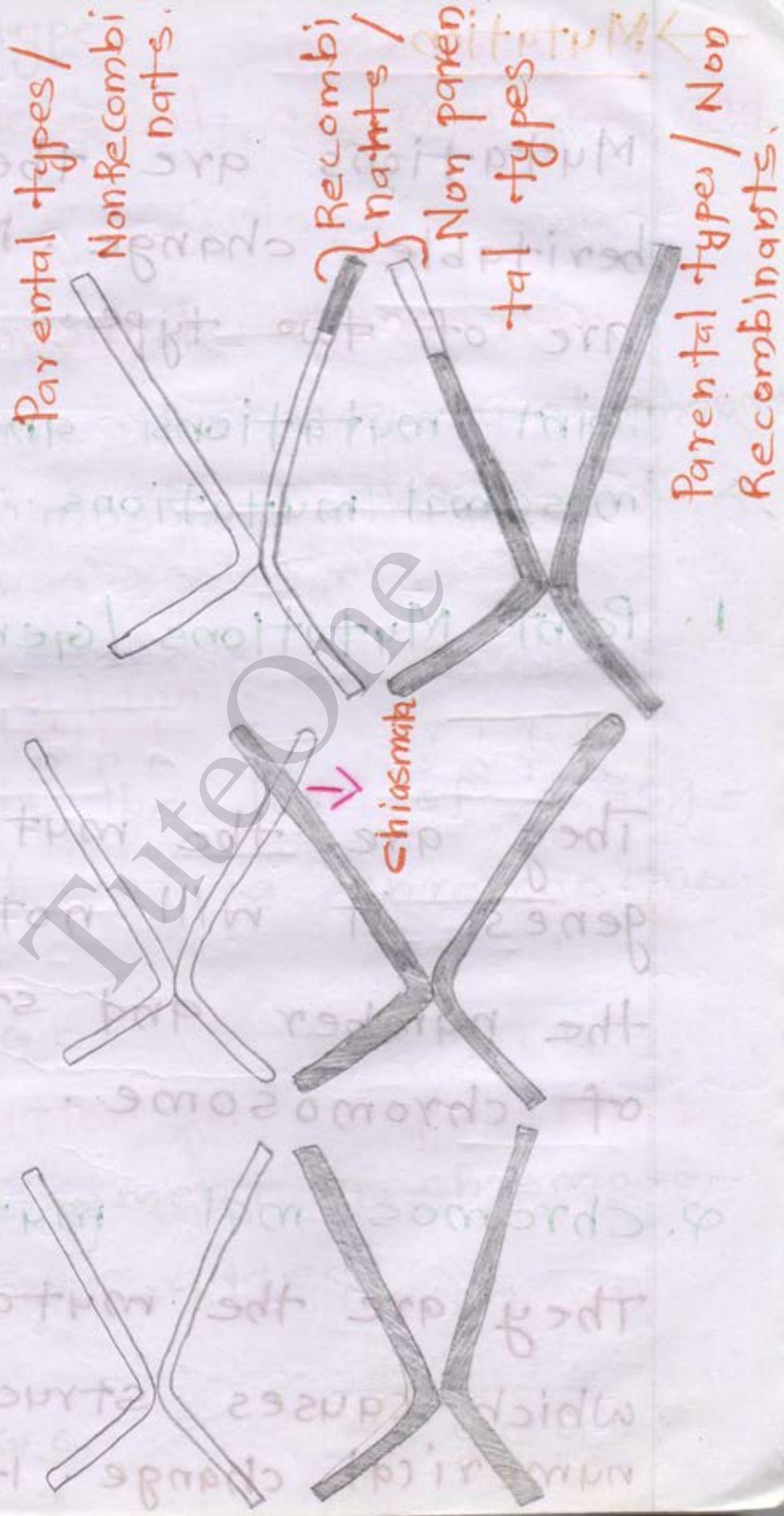
nation increases. As the

distance blw genes decreases

linkage increases and recom-

ination decreases.

Crossing over and Recombination



→ Mutation

Mutations are the sudden heritable change. Mutations are of two type.

Point mutations and chromosomal mutations.

1. Point Mutations / Gene mutation.



They are the mutations in genes it will not alter the number and structure of chromosome.

2. Chromosomal mutations.

They are the mutations which causes structural or numerical change. It is of

two types.

chromosomal aberrations and ploidy.

1. chromosomal aberration.

They are the structural changes in chromosomes. It is of 4 types.

(a). Deletion.

It is the loss of a segment of a chromosome.

ATGC \longrightarrow ATG

(b) Addition or Duplications.

A segment of chromosome is added.

ATGC \longrightarrow ATGC GC

(c) Inversion

A segment of chromosome is inverted.

ATGC → ATCG

(d) Translocation

It is the exchange of chromosomal segments.

ATGC AT34
1 2 3 4 1 2 GC

Q. Ploidy

It is the numerical changes in chromosomes. It is of two type.

(a) Euploidy

It is the change in chromosome number as sets of chromosomes.

$n - n \rightarrow$ Monoploidy / Haploid

$n + n \rightarrow$ Triploidy

$n + n \rightarrow$ Tetraploidy } Polyploidy

(b) Aneuploidy

It is the change in chromosome number as one or more chromosomes.

$n + 1 \rightarrow$ Trisomy

$2n - 1 \rightarrow$ Monosomy

$n - 0 \rightarrow$ Nullisomy.

→ Mechanism of Sex determination.

We have two types of chromosome Autosomes and Allosomes or sex chromosome.

Allosomes controls sex and Autosomes controls other body characters.

→ The mechanism of sex determination in human beings and in drosophila is XX-XY mechanism.

Male

$A+XY$

$A+X$

$A+Y$

Female

$A+XX$

$A+X$

Female $A+XX$

$A+XX$ male.

Males produces two types of gametes. So they are heterogametic and females are homogametic.

→ Mechanism of sex determination in insects is $XX-XO$ mechanism.

Male

$A+XO$

Female

$A+XX$

$A+X$

A

$A+X$

$A+XX$

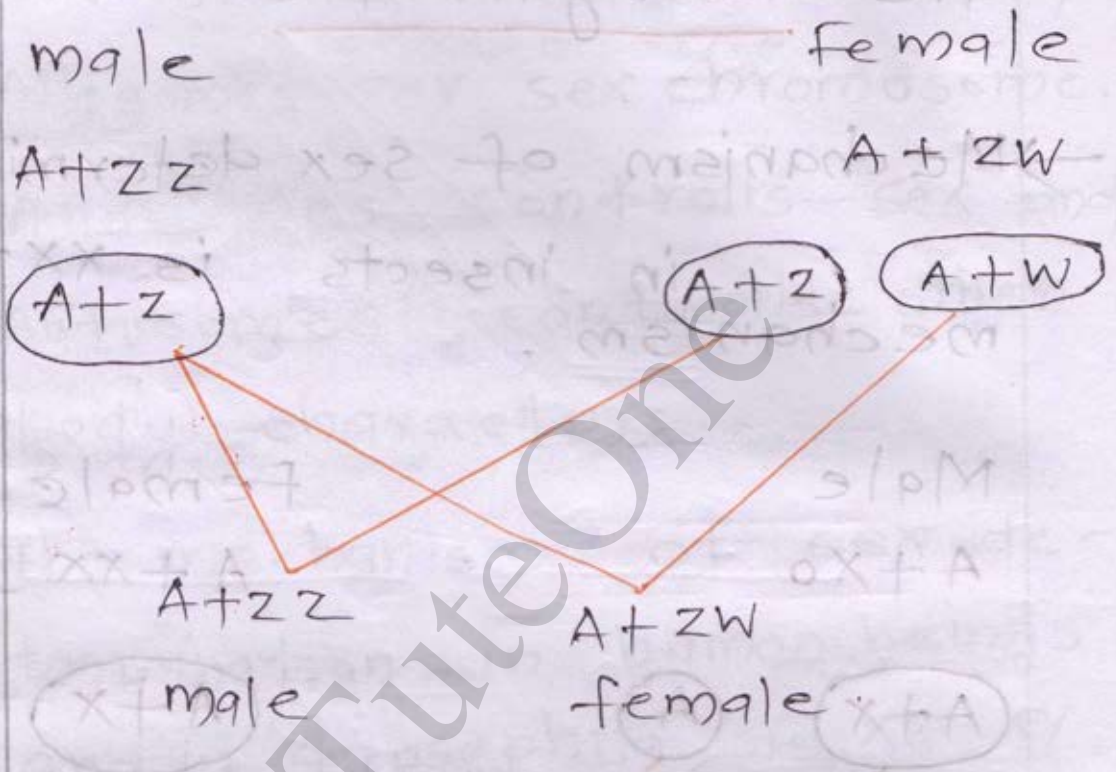
female

$A+XO$

Male.

Males are heterogametic and females are homogametic.

→ Mechanism of sex determination in Birds is ZZ-ZW mechanism.



females are heterogametic and males are homogametic.

Disorders.

Mendelian Disorders.

1. Hemophilia

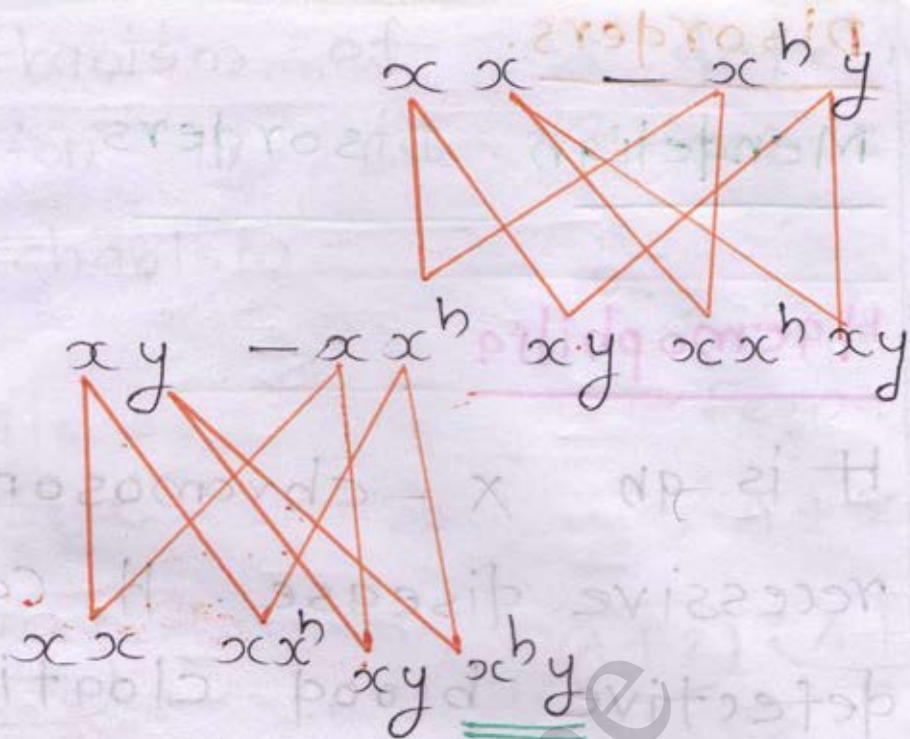
It is an x-chromosomal recessive disease. It causes defective blood clotting.

It is a royal disease. It is inherited from a person to its grandson, so known as criss-cross inheritance.

$x^h x^h \rightarrow$ Diseased female

$x^h x \rightarrow$ Carrier female

$x^h y \rightarrow$ Diseased male.



7. Sick leced anaemia

Hemoglobin has two polypeptide chains α and β . In the β chain at the 6th position amino acid is glutamic acid. In diseased persons it is replaced by Valine. So their RBC becomes in sickle shape and will not be able to transport oxygen.

$Hb^S Hb^S$ — Genotype of diseased person.

$Hb^S Hb$ — Genotype of carrier person.

3. Phenylketonuria

It is due to the absence of enzyme phenylalanine hydroxylase this converts phenylalanine to p -TH Tyrosine.

So the phenylalanine will be accumulated in body and converted into phenyl pyruvic acid. It will be deposited in brain causing brain damage and keton bodies are eliminated through urin.

chromosomal disorders.

1. Down's Syndrome.

It is due to 21^{th} trisomy.

They have 3 21^{th} chromosome. So they have 47

chromosomes with 45 Autosomes and 2 sex chromosomes.

It is known as Monosolism. They have mental retardation and congenital heart diseases.

2. Klinefelters Syndrome.

They have 44 Autosomes + XXY. So they have 47 chromosomes. So they are sterile males with female

characters (Gynaecomastia) .

3. Turners Syndrome

They have 44 Autosomes +
XO. They are sterile
females

→ Pedegree analysis

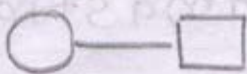
It is a chart to represent inheritance of a character or disease in human family.

○ → Female

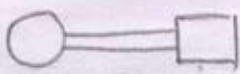
□ → male

◇ → Sex unspecified

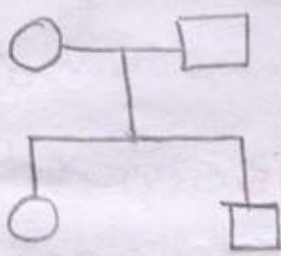
● → diseased



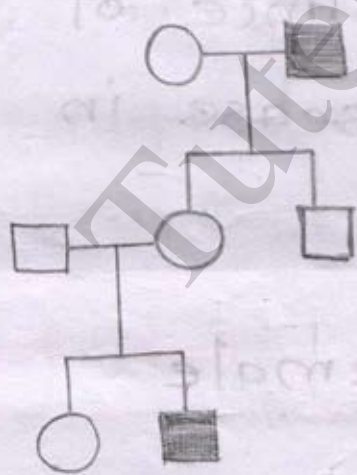
Marriage



Consanguineous
marriage



family

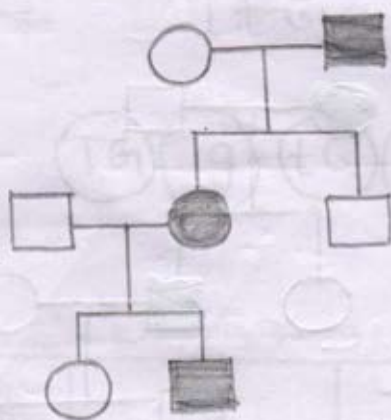


x — Recessive

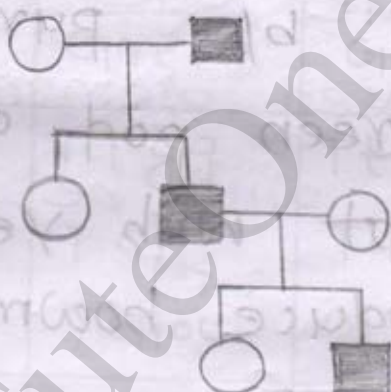
El parents

1 prozotvA

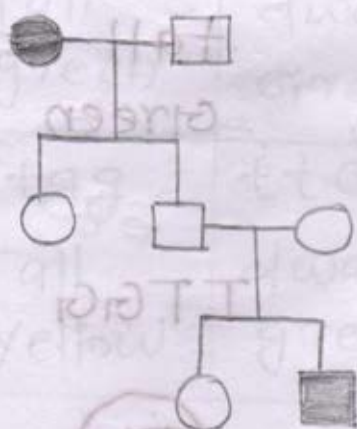
topalwop



X-Dominant

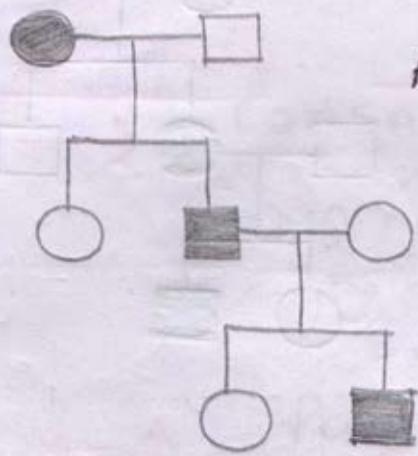


Y-chromo
Soma



Autosomal
Recessive.

Autosomal dominant



Autosomal dominant.

Q A cross b/w pure tall plant with green pod and dwarf pea plant with yellow pod - will produce how many shorts plants in F_2 .

An: Parents

tall
Green

X dwarf
Yellow

$TTGG$

$ttgg$

Gametes

TG

tg

F_1 generation.

$TtGg$ tall green

F₁ parent $TtGg \times TtGg$

Gametes $(TG)(Tg)(tG)(tg) \times (TG)(Tg)(tG)(tg)$

$\frac{\text{♀}}{\text{♂}}$	TG	Tg	tG	tg
TG	TTGG Tall Green	TTGg Tall Green	TtGG Tall Green	TtGg Tall green
Tg	TTGg Tall Green	TTgg Tall Yellow	TtGg Tall Green	Ttgg Tall Yellow
tG	TtGG Tall Green	TtGg Tall green	ttGG dwarf Green	ttGg dwarf green
tg	TtGg Tall Green	Ttgg Tall Yellow	ttGg dwarf green	ttgg dwarf Yellow.

phenotypic ratio - 9:3:3:1

Genotypic ratio - 1:2:1:2:4:2:1:2:1

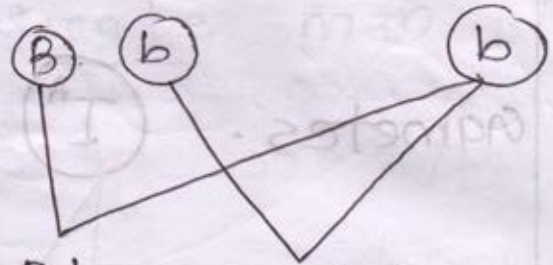
H will produce 4 short plants in F_2 .

Q Blue eye colour is recessive to brown eye colour. A brown eyed man whose mother was blue eyed marries a blue eyed woman. Their children will be

Ans: Brown — B
blue — b
mother — bb
man — Bb
woman — bb

Parent $Bb \times bb$

gametes



F₁ generation.

Bb

bb

Brown

blue.

1 : 1

Their children will be Brown or blue

Q A man of blood group A married a woman of blood group AB which type of their progeny indicates man is heterozygous.

An: Hetero
Parent.

$I^A i$

X

$I^A \cdot I^B$

Gametes.

I^A

i

I^A

I^B

F₁ gene
ration.

$I^A I^A$
A

$I^A I^B$
AB

$I^A i$
A

$I^B i$
B

Homo

Parent

$I^A I^A$

X

$I^A I^B$

Gamete

I^A

I^A

I^B

$I^A I^A$

$I^A I^B$

A

AB

B group blood grouped progeny indicate man is heterozygous.

Q A haemophilic man marries a woman carrier for haemophilia what would be the possibility to their daughter to be haemophilic.

Ans: Parents.

Father

Mother

$x^h y$

$x x^h$

gametes

x^h

y

x

x^h

$x x^h$

$x^h x^h$

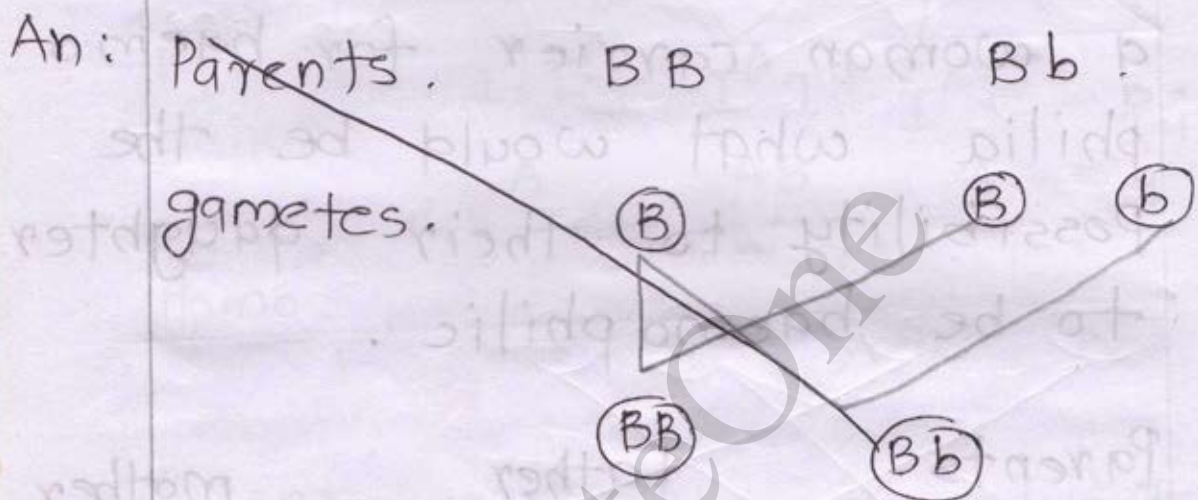
$x y$

$x^h y$

25%

The chance daughter should be haemophilic is 50%.

Q A brown eyed couple has a blue eyed child. find out the genotype of the couple.



man : BB or Bb

female : BB or Bb

P

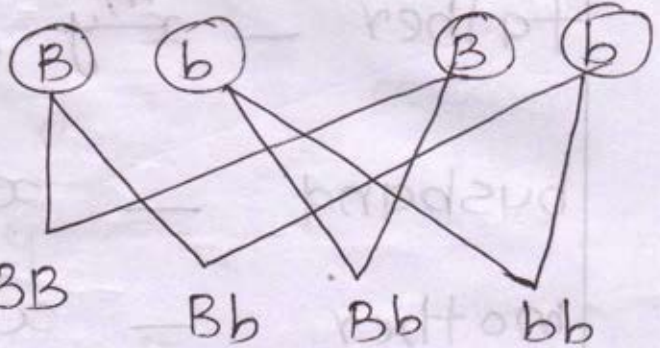
The genotype is Bb and Bb .

Parents I

Bb

Bb

gametes



F₁ generation

BB

Bb

Bb

bb

∴ The genotype of couple is

Bb

man

Bb

woman

Bb

Q. A husband and wife normal vision but fathers of both of them ^{were} ~~their~~ colour blind.

find out the possibility of their first daughter to be colour blind.

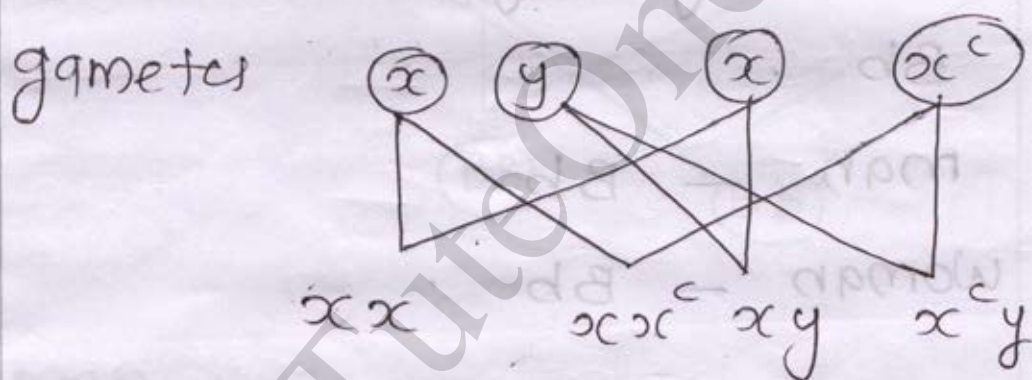
father — $x^h y$ $x^c y$

father — $x^h y$ $x^c y$

husband — $x y$

mother — $x x^c$

Parents $x y$ $x x^c$



The Possibility of their first daughter to be colourblind is 00 %.

Q A person whose father is colour blind marries a lady whose mother is daughter of colourblind man and woman.

Their children will be?

father — $x^c y$

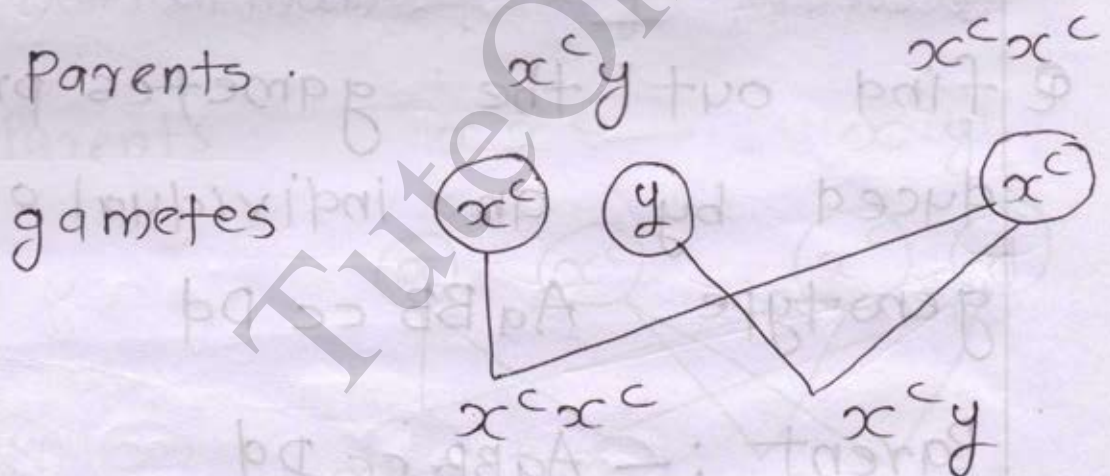
Son — $x y$

grand father — $x^c y$

grand mother — $x^c x^c$

Parents:

gametes



ladies mother — $x^c x^c$

lady — $x x$

Parents

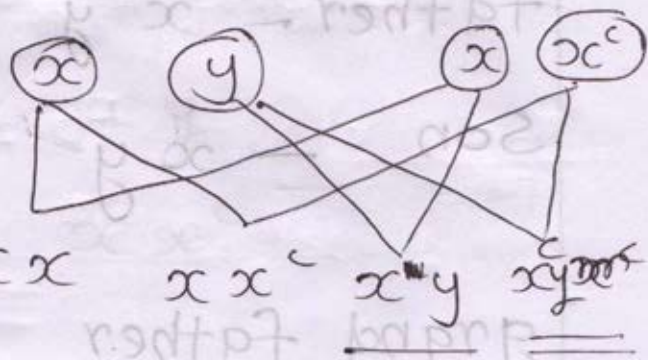
Son

lady

xy

xx^c

gametes



50% of their son.

Q find out the gametes produced by an individual of genotype $AaBBccDd$

Parent : — $AaBBccDd$

gametes $(ABcD)$ $(ABcd)$ $(aBcD)$ $(aBcd)$

no. of gametes = 2^n

n = heterogametic condition

Q A colour blind man has a colour blind sister and a normal brother. Find out the genotype of father and mother.

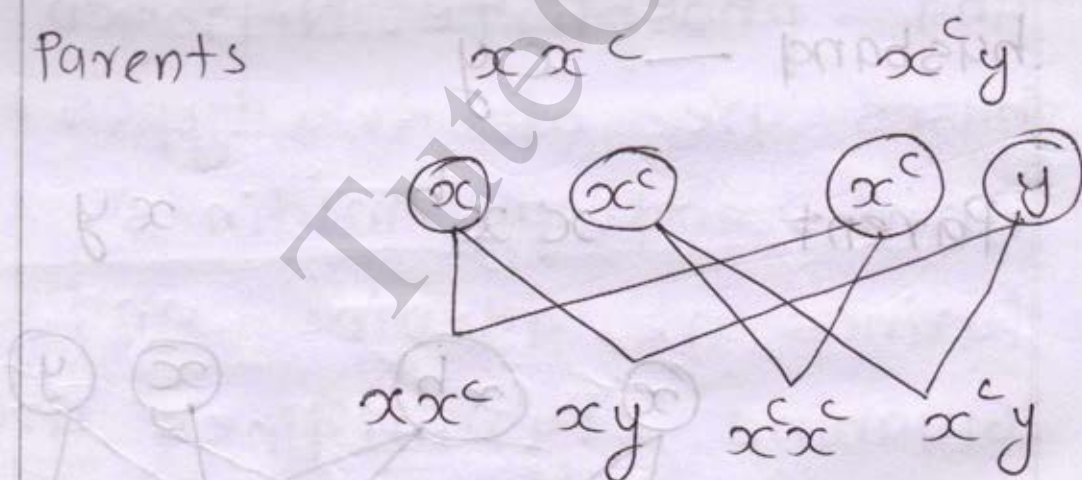
Ans:

man — $x^c y$

Sister — $x^c x^c$

normal man — $x y$

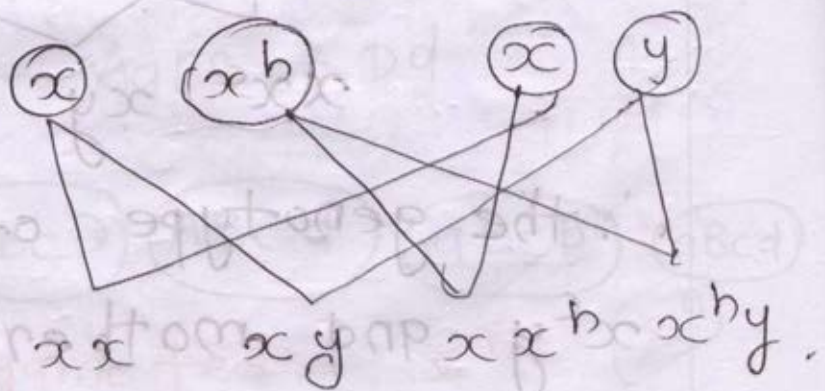
Parents



∴ the genotype of father is $x^c y$ and mother is $x x^c$.

Q A woman's father has haemophilia but her husband does not. what is the chance of their son to have the disease.

Ans: father — $x^h y$ $x x$
 woman — $x x^h$
 husband — $x y$
 Parent $x x^h$ $x y$



Their son has to be disease 50%.

50% chance

INHERITANCE

← Structure of DNA

of the DNA is formed of nitrogenous bases, phosphate sugar and phosphate. Nitrogenous bases are of two types purines and pyrimidines. Purines are adenine and guanine. Pyrimidines are cytosine, thymine and uracil. When a nitrogenous base is joined with a phosphate sugar by glycosidic bond, it forms