Module-05

Genetics

Date: / /
Page No.:

Genetics :- Genetics is a branch of biology concerned with the study of genes, genetic variation.

Father of Genetics :- Gregor Johann Mende

↳ Conducted hybridisation experiments on garden pea (Pisum sativum) for 7-years and gave the laws of inheritance in living organisms. 14 - true breeding pea plant.

⇒ Garden Pea Selection Reasons :-

① Available in many varieties on a large scale to observe alternate traits.

② Peas are self-pollinated and can be cross-pollinated.

③ Short life span, so several generation can studied within a short period.

④ Easily be raised, maintained and handled

⑤ Many varieties are available with distinct characteristics which plants provide many easily detectable contrasting characters.

Date: / /
Page No.:

Mendel's Law

Mendel's laws of inheritance are based on monohybrid crosses.

(1) Principle of Dominance

This law states that when two alternative forms of a trait are present in an organism, only one factor express itself in f_1- progeny and is called dominant, while the other that remains masked is called Recessive.

↳ f_1- generation ⟶ only one character express.

↳ f_2- generation ⟶ Both characters are expressed.

Punnett squares

T — tall plants.
t - short plants

f_1- Generation.

	T	T
t	Tt	Tt
t	Tt	Tt.

⇓

All are tall plants.

f_2- Generation.

	T	t.
T	TT	Tt.
t.	Tt	tt.

Tall → TT, Tt, Tt.
short → tt.

* Phenotype :- 3:1.
* Genotype :- 1:2:1

② Law of Segregation (1st Law).

↳ This law states that the alleles do not show any blending (Mix-up) and both the characters are recovered in f_2 - generation.

↳ The gametes are pure for a character.

↳ The parents contain two Alleles during gamete formation.

↳ The alleles of a pair segregate from each other such that a gamete receives only one of the two alleles.

③ Law of Independent Assortment (2nd Law)

↳ This law states that when two pairs of traits are combined in a hybrid, segregation of one pair of character is independent of the other pair of characters at the time of gamete formation.

↳ Based on the results of dihybrid crosses, where inheritance of two traits were considered simultaneously.

Date: / /
Page No.:

↳ Mendel performed di-hybrid cross in pea plants that were true breeding for two traits.

eg - Round seeds and yellow seed color was cross-pollinated with a plant that had wrinkled-seed and green seed color.

↳ In this cross — RR - Round seeds and yellow seed (YY) are dominant.

↳ Wrinkled seeds (rr) and green seed (yy) are Recessive.

↳ The resulting offspring f_1 generation were all heterozygous for round seed and yellow color (Rr Yy).

↳ Mendal allowed f_1 plants to self-pollinate, thus in f_2 generation he noticed a ratio of 9:3:3:1 in the phenotypes.

Date: / /
Page No.:

Allele

↳ An allele is an alternative form of a gene (one member of a pair).

↳ Location:- Specific position on a specific chromosomes.

↳ Allele determine distinct traits that can be passed from parents to child through sexual reproduction.

↳ The process by which alleles are transmitted was discovered by Medel's.

↳ Diploid organisms typically have two alleles for a trait.

↳ Homozygous:- Alleles pairs are same.

↳ Heterozygous:- Alleles pairs are different.

↳ Dominant alleles is expressed and the recessive allele is masked. This is called complete Dominance.

↳ In Heterozygous relationships where neither allele is dominant but both are completely expressed is called co-dominant. eg AB blood type.

(P.T.O)

Date: / /
Page No.:

↳ When one allele is not completely dominant over the other, the alleles are said to be Incomplete Dominance.
eg— Pink flower of tulip.

⇒ Multiple Allele:-

↳ Most genes exist in two allele forms, some have multiple alleles for a trait.
eg - Human ABO - blood type.

↳ Human blood type is determined by the presence or absence of certain identifiers, called Antigens.
eg - blood type 'A' — A antigens.
Blood type 'B' — B antigens.
Blood type 'O' — No antigens.

↳ ABO - blood type exists as three alleles as (I^A, I^B, I^O).

↳ There are four phenotypes (A, B, AB or O) and six possible genotype for human blood.

Blood Group.	Genotype
A	(I^A, I^A) or (I^A, I^O).
B	(I^B, I^B) or (I^B, I^O)
AB	(I^A, I^B)

Gene mapping :-

↳ Gene mapping is the process of establishing the locations of genes on the chromosomes.

↳ Give informations about genetic diseases.

↳ Such maps indicate the position of genes in the genome and also distance btw them.

↳ Gene that are on the same chromasom are said to be linked and the distance b/w these genes is called a linkage distance.

↳ Genes can be viewed as one special type of genetic markers

↳ There are two type of gene/Genome mapping —

(i) Genetic mapping.
(ii) physical mapping.

Eg- DNA markers:-

(i) RFLP :- Restriction fragment length polymorphism.

(ii) SSLP :- Simple sequence length polymorphism.

(iii) SNP :- Single nucleotide polymorphism.

Date: / /
Page No.

(1) Genetic mapping :-

↳ Based on the use of genetic technique to construct maps.

↳ These maps show the position of genes and other sequence ~~of genes~~ features on a genome.

↳ Also helps to determine the relative position b/w two genes on a chromosomes.

↳ Genetic maps are guiding Scientists to many genes related diseases. eg — Asthama, heart disease, diabetes, cancer, psychiatric conditions ... etc.

(2) Physical mapping :-

↳ uses molecular biology technique to examine DNA molecules directly.

↳ Based on these techniques map construction is done.

↳ These maps shows the position of sequence features including genes.

* All maps are based on phenotype of the organisms.

Date: / /
Page No.:

Gene Interaction.

The influence of allele and non-allele on normal phenotypic expression of genes.

⇒ Types of gene Interaction.

① Allelic / non-epistatic gene Interaction.
② Non-allelic / epistatic gene Interaction.

① Allelic / non-epistatic Gene Interation:-

When 2-allele (present on same gene locus on homologous chromosome) of a gene, interact in such a way to produce a phenotype differ from typical dominant-recessive phenotype.

Eg - incomplete / co-dominance, multiple allele.
~~dominance~~.

② Non-allelic / epistatic gene Ineraction:-

When 2- or more independent gene present on same or different chromosome, interact to produce a different expression.

eg - Epistasis, complementary gene, supplementary gene, etc.

Date: / /
Page No.:

(1) Complementary gene :-

(1) Two pair of non-allelic genes are essential in dominant form to produce a particular character.

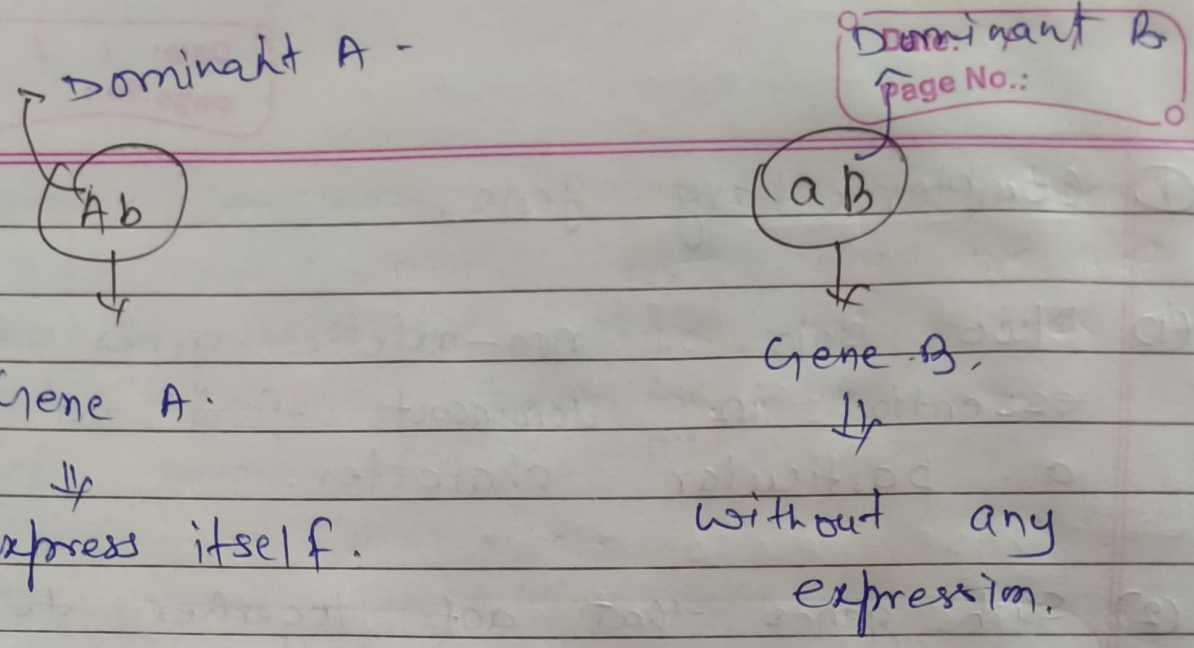
(2) Such genes that act together to produce an effect that neither can produce, its effect separately are called complementary genes.

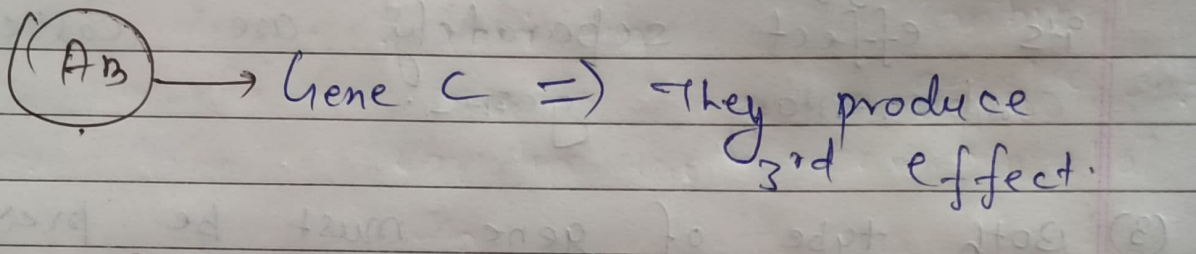
(3) Both type of gene must be present in dominant form.

eg - flower color in garden pea.
↳ Two pea plants — white color.
↳ F_1 generation $\xrightarrow[\text{pollination}]{\text{Cross}}$ purple color.
↳ F_2 generation $\xrightarrow[\text{pollination}]{\text{Cross}}$ 9P : 7W.

(2) Supplementary Gene Interation :-

Supplementary genes are pair of non-allelic genes. One of which produce its effects independently in the dominant state while the dominant ~~state~~ allele of the second gene is with any independent effect but is able to modify the effect of the former to produce a new traits.





eg – (1) coat colour in Mice.
(i) Dominant (B) allele – Black.
(ii) Dominant (A) allele – Albino
(iii) Both dominant (AB) allele – Agouti
(iv) Both recessive (ab) allele – Albino.

phenotype ration → F_1 – All agouti.
→ F_2 – 9a : 3b : 4albino.

(3) Epistasis :–

Suppression of the effect of a gene by another gene. One gene completely masks another gene.

eg - coat color in mice.

Date: / /
Page No.:

Cells divide And Reproduce in two ways.—
① Meiosis ② Mitosis.

	① Meiosis	② Mitosis
*Defination:-	Types of cellular reproduction in which the number of chromosomes are reduced by half through the seperation of homologous chromosomes, producing two haploid cells.	Process of asexual reproduction in which the cell divides in two producing a replica, with an equal number of chromosomes in each resulting diploid cell.
*Reproduction:-	sexual	Asexual.
Occurs:-	Humans, animals, plants.	All organisms.
*Chromosomes number:-	Reduced by half.	Remains the same.
* crossing over:-	Yes, mixing of chromosomes occuss.	No, crossing over can't occurs.
* creates:-	sex cells only	Makes everything other than sex cells.
* functions:-	Genetic diversity through sexual reproduction.	cellular reproduction and general growth and repair of the body.
* Steps:-	Meiosis 1. Meiosis 2.	prophase Metaphase Anaphase Telophase,

Date: / /
Page No.:

Variation :- The individuals of a species may look similar but they are not usually identical, these differences are called variation.

Varriation.

- Genetic variation / Inherited variation.
  - ↓
  - Variation due to genetic cause.
  - eg - Eye color, Hair color, Skin color.
- Environmental variation
  - ↓
  - variation due to environmental conditions.
  - eg - Climate, diet, culture, Lifestyle.
- Combined genetic and Environmental
  - ↓
  - Variation of combinations due to boths.
  - eg - twins. growth...

Date: / /
Page No.:

Concepts of Recessiveness And Dominance:-

↳ Gene contain specific information.
↳ Everyone inherits two copies of each gene from their parents; and copies are different or the same.
↳ If the copies are different, then one is going to mask the effects of the other.
↳ The gene that trumps over the other is usually known as the Dominant gene. and the weaker gene is known as Recessive.
↳ In the presence of a dominant gene, a recessive gene will not show its traits.
↳ When two recessive genes are paired their traits will be visible.
↳ Dominant genes are also the one whose traits are visible in the offspring.
↳ For recessive traits to be visible both parents have to carry that recessive gene singly or in pair.
↳ Genes are simply an instruction manual used to make certain protein. Protein is actually responsible for the traits that are presented physically.
↳ Two copies of each gene (from both parents).

(P.T.O)

Date: / /
Page No.:

↳ The combination of these two genes will cause the formation of a slightly different proteins.

↳ When a gene makes a protein that is functional, then that gene is dominant.

↳ If the protein formed is broken then the gene that has formed is recessive.

⇒ Genotype

The genetic makeup of an organism. The gene combination of an organisms.

⇒ Phenotype

The physical characteristics of an organism. They m organism look

Date: / /
Page No.:

Single Gene disorders in Humans:-

↳ Single gene disorders are caused by DNA changes in one particular gene and often have predictable inheritance patterns.

↳ 10,000 human disorders are caused by a change, known as mutation in a single gene. or single gene disorders.

↳ The mutated version of the gene ~~rep~~ responsible for the disorder is known as a mutant or disease.

⇒ Gene mutation in autosomes:-

① Recessively inherited trails:- These are caused by recessive genes in homozygous condition. eg—

(i) ~~Alkap~~ Alkaptonuria:- It is caused by disorder in single gene. The symptom may include blackening of urine on exposure to O_2 and darkening of cartilage.

(ii) Albinism:- It is caused by lack of pigment melanin in skin, hair and iris of eye. It is caused by absence of enzyme tyrosinase which produce melanin. It is seen when both the alleles of gene are recessive.

(iii) Sickle cell Anemia :- It is autosomal recessive disease so it is transmitted from parents to offspring when both parents are carrier (Heterozygote) for the gene.

The disease is controlled by single pairs of alleles ~~Hbf~~ Hb^A and Hb^S.

This disease is caused when glutamic acid is replaced by valine.

(iv) Thalassemia :- It is autosomal recessive disease. It is caused by defect in synthesis of globin polypeptide in RBC resulting in sever anaemia.

X- linked Inheritance

↳ X-linked inheritance means that the gene causing the trait or the disorder is located on the X- chromosomes.

* females — XX * Males — XY

↳ X-linked Recessive genes are only expressed in female.

↳ for males, there needs to be only one copy of an X-linked recessive gene in order for the trait.

(P.T.O)

Date: / /
Page No.:

Hemophilia :- A disorder in which blood desn't clot normally.

↳ Hemophilia is an inherited genetic condition.

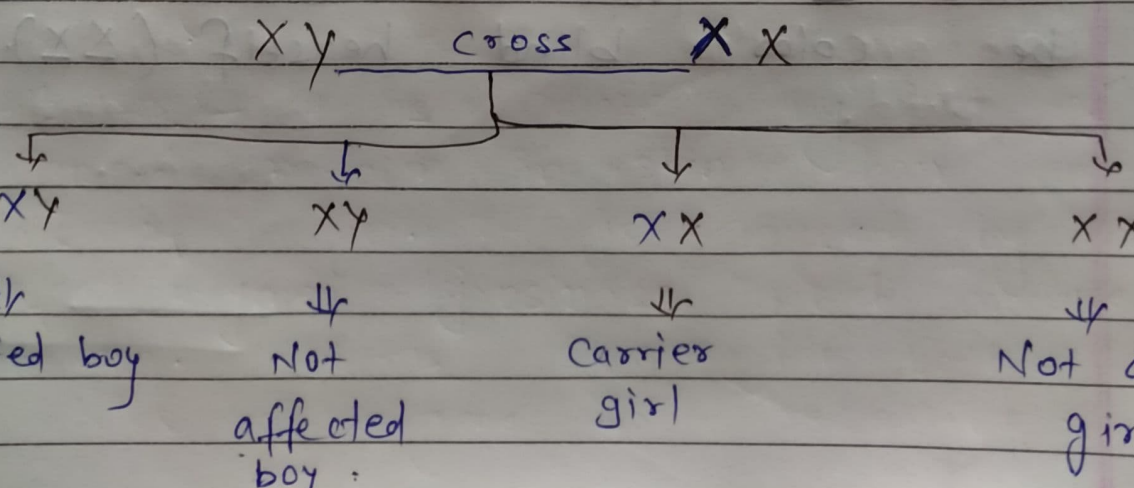
↳ These gener are located on x-chromosomes. making hemophilia x-linked recessive diseases.

↳ Father can't pass the diseases to their sons.

↳ If male gets the x-chromosome with the altered gene from his mother, he'll have hemophilia.

↳ 50% chance of passing that gene to her children.

↳ A female who has the altered gene on one of her x-chromosome is typically called a Carrier. This means. She may pass the disease to her children but she doesn't have the diseases herself.



Date: / /
Page No.:

Color blindness :- Not able to recognise green and red color.

* Normal X-chromosome — X
* Color blind x-chromosome — $\underline{X}$

↳ Color blind gene is carried on one of the X-chromosome. since men have only one X-chromosome.
If he ~~carries~~ color blind ~~color~~ then ($X\underline{Y}$).

↳ A women can be :-

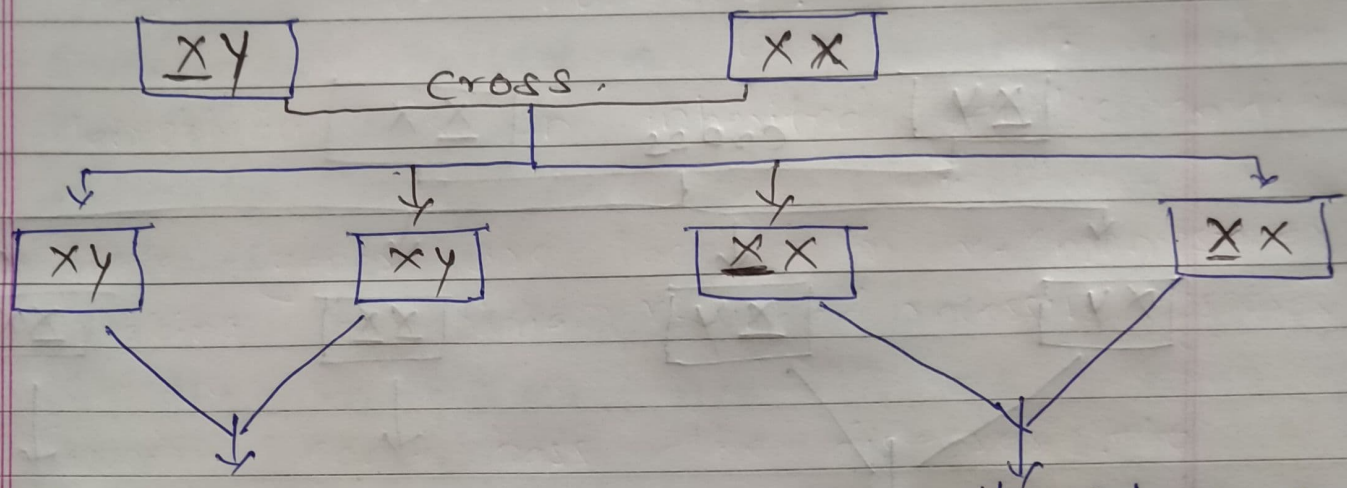
(i) Two normal X-chromosomes, so that she will not be color blind or be a carrier (XX).

(ii) One normal X and one color blind carrying $\underline{X}$ chromosome, in which case she will be a carrier ($X\underline{X}$).

(iii) She will inherit a color blind $\underline{X}$ from her father and a color blind $\underline{X}$ from her mother and be color blind herself ($\underline{X}\underline{X}$).

Date: / /
Page No.:

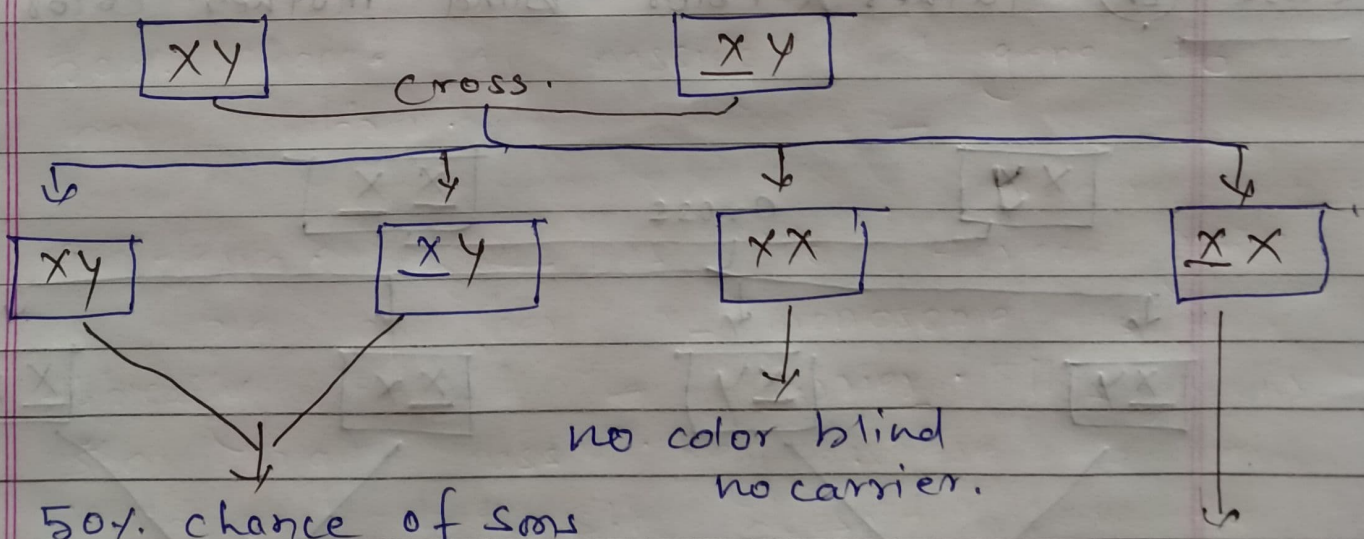
Case (I) father color blind normal mother.



both son are not color blind.

both daughters are color blind carried.

Case (II) color blind mother & Normal father.



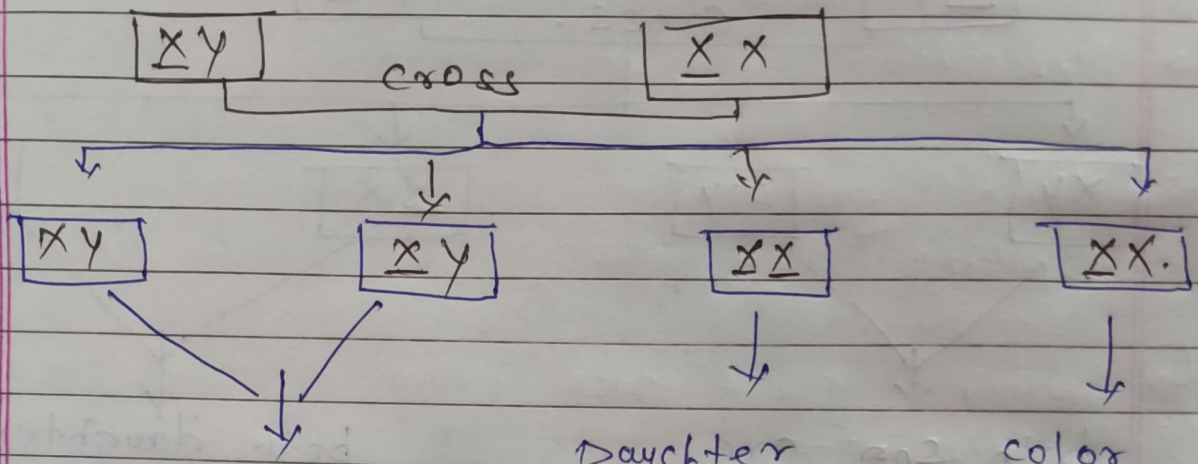
50% chance of sons being color blind.

no color blind no carrier.

color blind carrier.

Date: / /
Page No.:

Case-III Both parents are color blind.

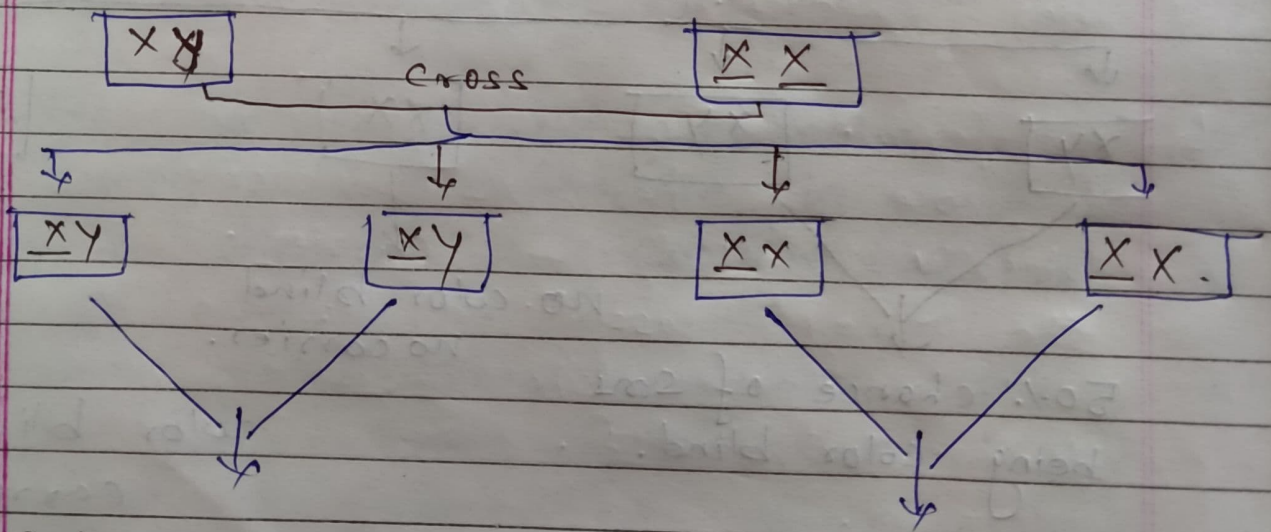


50% chance of sons being color blind.

Daughter color blind.

color blind carrier.

Case-IV father not color blind mother color blind.



Both are color Blind.

Both are color blind gene carrier.

Complementation Test.

Sometimes called a cis-trans test. It can be used to test whether the mutations in two strains are in different genes.

complementation will not occur if the mutations are in the same gene.

It is test for determining whether two mutations associated with a specific phenotype represent two different forms of the same gene (alleles) or are variations of two different genes.