UNIT VII- Genetics and Evolution

CHAPTER: 5 – Principles of Inheritance and variation

<u>Introduction:→</u> Gregor John Medel (1822-186) was first to study about the mode of transmission one generation to another. He began his famous experiment on pea (Pissum sativum). He conducted hybridization experiments on pea for seven years (1856-1863) and formulates the laws of Inheritance in living organisms. For the act of this, he is regard as the father of Genetics.

Genetics: The branch of science which deals with the mechanism of heredity transmission and causes of variation in living being is known as genetics.

Inheritance or Heredity: It is the process of transmission of characters from one generation to another. The characters which are transmitted from parents to offspring is known as heredity trait.

Variation: The slide differences shown by individuals of a species and also by the offsprings of the same parent are called variation.

Mendel' Experiment: Mendel select Pea plant (Pissum sativum) for his Experiment for the following reasons

i) Pea plant is a self-pollinating plant but cross pollination can be achieved.
ii) It is easy to cultivate.
iii) It has a no. of inherent properties.
iv) It is a hermaphrodite of bisexual plant.
v) It shows self-fertilization and thus plants are homozygous. It is therefore easy to get pure lines for several generation.

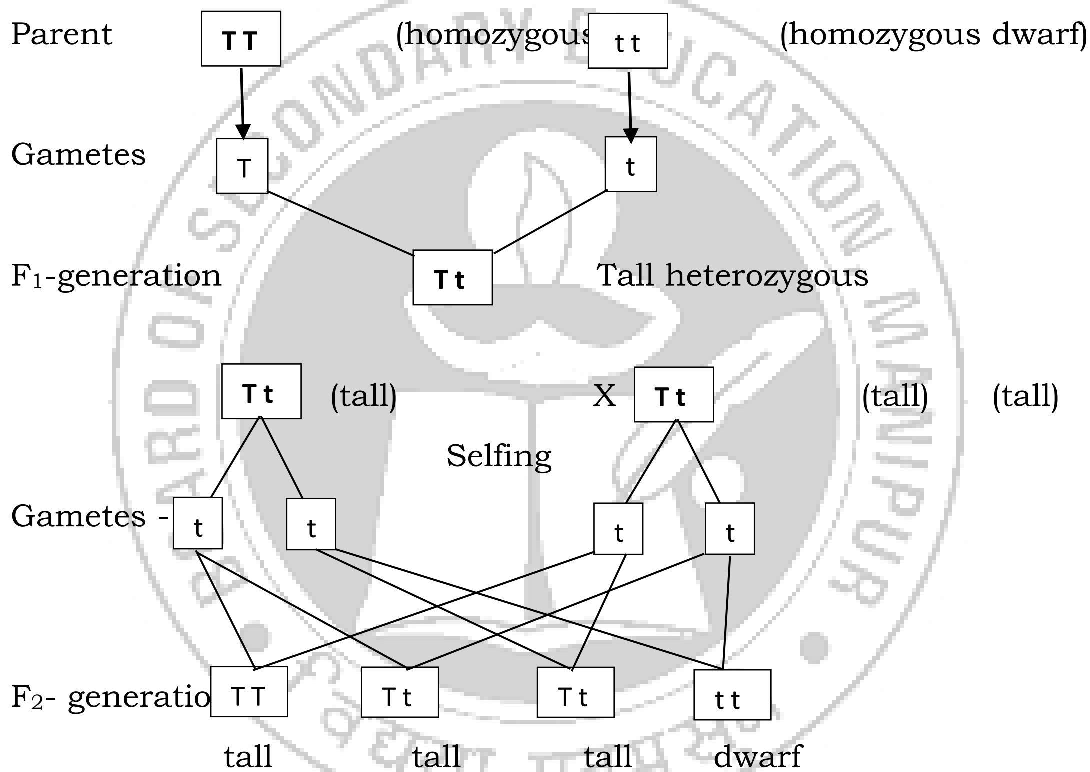
List of seven pairs of contrasting trait in Pea plant.

Sl.No	Characters	Dominant trait	Recessive trait
1.	Stem length	Tall	Dwarf
2.	Seed shape	Smooth (round)	Wrinkled
3.	Seed colour	Yellow	Green
4.	Flower colour	Violet	White
5.	Pod shape	Inflated	Constricted

6.	Flower position	Axial	Terminal
7.	Pod colour	Green	yellow

Mendel's Experimental Work: Mendel conducted artificial or cross pollination experiment using tree breeding lines. A tree breeding line is one that has undergone continuous self-pollination and shows stable trait inheritance for several generation.

Monohybrid cross:-

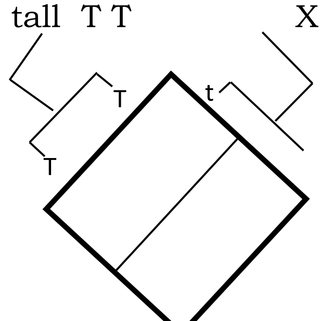


Phenotype Ratio → tall (3) : dwarf (1)

Genotypye Ratio → T T (1) : T t(2) : t t (1)

Punnett Square: Graphical representation to calculate the probability of all possible genotypes of offsprings in a genetic cross.

Pure tall T T X t t pure dwarf



Gametes - Tt t

F_1- generation Tt Tt

Tt all hybrids are tall.

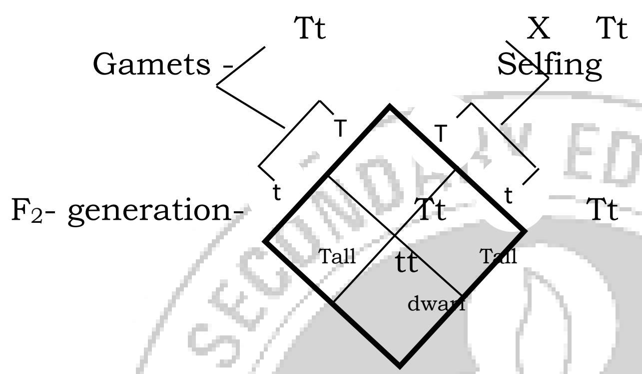


Fig:→ A Punnett Square of a typical Monohybrid cross between tall pea plant and dwarf pea plant.

i) **Monohybrid cross**
ii) **Dihybrid cross**

Monohybrid cross: When we consider only one pair of contrasting character in a cross, then it is known as monohybrid cross.

Eg. The cross between tall and dwarf pea plant.

Dihybrid cross: The cross in which the inheritance of two pairs of contrasting characters is considered at a time, it is called dihybrid cross.

Eg. The cross between Yellow, Round seed with Green wrinkled seed.

Explanation of Mohybrid cross:

Mendel crossed tall and dwarf pea plant to study the inheritance of one gene. He collected the seeds produces as a result of this cross and grew them to generate plants of the first hybrid generation called F_1 progeny.

The F_1- progeny were tall, none was dwarf. He again self-pollinated the tall F_1- plants and found that $\frac{1}{4}^{th}$ of the F_2- plants were dwarf, while $\frac{3}{4}^{th}$ of the F_2- plants were tall.

Thus only one of the parental traits were expresses in the proportion 3:1. The contrasting traits did not show any bleeding at either F_1 or F_2 stage.

Mendel's Experimental Result:

Gene :-> It is the functional unit of heredity material. or It is the particular segment of DNA molecule which determine the hereditary of a particular trait. It is equivalent to 'factor'.

Allele :-> Two or more alternative forms of gene are called alleles.GE

Genotype :-> The genetic make up or genetic constitution of an individual is known as genotype.

Eg. Red flower } Genotype. = (RR)

White flower=(rr)

Phenotype :-> The external appearance of an organism for a particular contrasting character is called phenotype.

Eg. Tallness, dwarfness.

Homozygous :-> Identical alleles are present for a particular trait, then it is called homozygous.

Eg. Tall } Homozygous (TT)

Dwarf (tt)

Heterozygous :-> If different alleles are present for a particular trait, that it is called heterozygous.

Eg. Tall } Heterozygous (Tt)

Red (Rr)

Dominance:-> It is a phenomenon in which one member of a pair of allelic gene express itself as whole (complete)or in part (incomplete).

Eg.

Tt – tall (Complete Dominance)

Rr- pink (Incomplete Dominance)

Recessive :-> A phenomenon in which out of two contrasting trait, when a single trait fail to express itself in the successive generation, then it is called recessive.

<u>LAWS OF INHERITANCE</u>

i) **Law of Dominance**
ii) **Law of Segregation**
iii) **Law of Independent Assortment.**

i) **Law of Dominance :->** In a monohybrid cross, only one gene out of the allelic pair express itself as whole in F_1-hybrid while the other does not shown its effect. The express gene is called dominant gene and hidden gene is called recessive gene.

Dihybrid Cross showing law of Independent Assortment:

Yellow Rounded seed X Green wrinkled seed

Parent → YYRR yyrr

Gametes → YR yr

F_1-hybrid - YyRr - yellow round seed

YyRr X 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 round	YyRr yellow round	Yyrr yellow wrinkled
yR	YyRR yellow	YyRr yellow	yyRR green	yyRr green

F_2-genaration→

	round	round	round	round
yr	Yyrr yellow wrinkled	Yyrr yellow wrinkled	yyRr green round	yyrr green wrinkled

Phenotypic Ratio- yellow round : yellow wrinkled : green round : green wrinkled

9 : 3: 3: 1

Thus law of dominance is used to explain the expression of only one of the parental character in a monohybrid cross in F_1 – and expression of both the characters in F_2.

ii) **Law of Segregation:** "Allelic pair segregate or separate at the level of gametogenesis". The paired condition of gametes during fertilization and both the characters are recovered in F_2-generation.

iii) **Law of Independent Assortment:** During inheritance of two or more pair of contrasting characters, the distribution of gene during gamete formation and in the progeny of subsequent generation is independent with each other and they assort themselves independently at random fusion.

Mendel cross a variety of pea having yellow round (YYRR) with other having green wrinkled (yyrr) seed. The F_1-hybrids on selfing (pollinating among themselves) produces four kinds of plants in F_2-generation. They were in the ratio of 9 (yellow round): 3 (yellow wrinkled): 3 (green roud): 1(green wrinkled).

Thus Rhenotypic Ratio -> 9:3:3:1

BACK CROSS AND TEST CROSS:

Back Cross :-> When F_1-hybrids are crossed with one of the two parents from which they are derived, these such cross is called back cross. When

F_1-hybrid is back cross with the dominant parent, no recessive individuals are obtained in the progeny.

TT (dominant parent) X (Tt)-F_1

Gamete – T T T t

F_2 -- TT Tt TT Tt

tall tall tall tall

$\frac{1}{2}$ TT : $\frac{1}{2}$ Tt
tall tall

Fig. Back Cross

(F_1) Tt X tt (Recessive parent)

T t t t

Tt Tt tt tt
tall tall dwarf dwarf

$\frac{1}{2}$ Tt : $\frac{1}{2}$ tt
tall dwarf

Fig. Test Cross

TEST CROSS :-> When F_1-hybrid is back cross with the recessive parent, equal of dominant and such type of back cross is known as test cross.

It is used to test an individual, either homozygous or heterozygous.

<u>Incomplete Dominance:</u>

Parent → RR (Red) X rr (White)

Gamete → R r

F_1 – Rr

F_2- Rr Selfing X Rr

Gametes- R r R r

F_2 - RR Rr Rr rr
Red Pink Pink White

Genotypic Ratio → RR(1) : Rr(2) : rr(1)
Phenotypic Ratio→ Red(1) : Pink(2) : White(1)

F_1-G:-> Result of monohybrid cross is Snapdragon where 1 allele is incompletely. Dominant over the other allele.

DEVIATIONS FROM MENDELISM:

i) Incomplete Dominance :-> The law's dominance does not occur universally. In certain case, they dominant recessive gene could not dominate completely or express itself as a whole instead they interact to produce an intermediate character in F_1- generation is known as incomplete dominance.

Eg. Snapdragon

ii) Co-Dominance :-> In co-dominance, the dominant and recessive gene contribute equally to the phenotypic character and they occur side by side in F_1-hybrid to give a mix character.

Eg. (i) Blood group AB :-> In blood group 'AB' both the alleles I^A and I^B could not dominant but they contribute equally in the phenotype of human blood group.
i.e. AB Blood group = (I^AI^B)genotype.

iii) Multiple Allele :-> It is a set of three, four or more alternative forms of a gene which have arisen as a result of mutation of the normal gene and which occupy the same locus in the homologous chromosome.

Eg. ABO blood groups are controlled by the gee 'I'. The gene 'I' has three alleles I^A, I^B and I^o.

Multiple Allele

I^A —— I^AI^A —— A

I —— I^B —— I^BI^B —— B

Immunogen gene —— I^AI^B —— AB

—— I^O —— I^OI^O —— O

'I'- gene exhibit in 3 allelic form | allelic pair | Blood group

Blood group Inheritance :->

Case I — Mother Blood group A; Father Blood group B

Genotype: I^AI^O × I^BI^O

Gametes: I^A, I^O × I^B, I^O

F1-generation: I^AI^B, I^AI^O, I^BI^O, I^OI^O

Blood Group: AB, A, B, O

Case II:-> Parents between blood group AB and O.

Parents- I^AI^B × I^OI^O

Gametes - I^A, I^B × I^O, I^O

Genotype - I^AI^O, I^AI^O, I^BI^O, I^BI^O

Blood Group: A, A, B, B

Chromosomal theory of Inheritance: [Sutton and Beveri]

1) The genes are located in the chromosome and each chromosome bears many genes.
2) The genes located on the same chromosome tends to inherit to together. The inheritance of gene together and to retain their parental combination is the offspring is known as linkage.

3) The somatic cells of an organism have homologous pair of chromosome each receiving from maternal and paternal parent.
4) Each chromosome contain numerous genes which help the organism to develop from zygote.
5) Each chromosome thus maintain its requireness individuality and its continuity throughout the life cycle of an organism.

REASONS FOR SELECTING DROSOPHILA FOR STUDY GENETICS:

Major worked on fruit fly (Drosophila melanogaster)

1) Its life cycle is very short (10-12 days).
2) It can be cultures throughout the year in small room.
3) The culture technique for rearing is also very simple and easy to handle.
4) It is highly proliferative for which large number of progenies can be quickly obtained in each breed.

LINKAGE AND RECOMBINATION

Linkage - Morgan, 1910

Morgan termed the physical association of two genes as linkage. All the genes present on the same chromosome are said to be linked genes and should be inherited together.

Theory of linkage :->

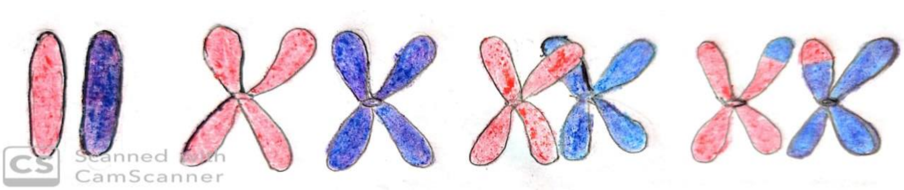
1) Linked genes occur in the same chromosome.
2) They lie in a linear sequence in the chromosome.
3) There is tendency to maintain the parental combination of genes.
4) Strength of the linkage between two gene is inversely proportioned to the distance between the two.

Linkage is of two type- i) Complete linkage ii) Incomplete linkage.

Complete linkage:- The genes located in same chromosome do not separate and are inherited together due to absence of crossing over. Thus combination of parental type trait is inherited.

Incomplete linkage:- Genes present in the same chromosome have tendency to separate due to crossing over and produced recombinant progeny besides parental type.

CROSSING OVER:



i) Synapsis ii) Tetrad formation iii) Crossing over iv) Disjunction

Crossing over is the exchange of genetic materials between non-sister chromatids of homologous chromosomes.

Mechanism- i) Synapsis:- During prophase-I of meiosis pairing f homologous chromosome take place.

ii) Tetrad formation:-They duplicated or splited lengthwise in sister chromatids and form tetrad.

iii) Crossing over:- In pachytene stage, non-sister chromatids come in contact at the point chiasmata and at this point, the chromatid segments break and rejoined after exchanging their segments. This phenomenon is known as crossing over.

iv) Disjunction:- As a result of crossing over, new combinations are produced and homologous chromosome move apart.

Significance: It bring variation and new combination (recombination) of character in the offspring.

Recombination:- A new grouping of genes or new combination of characters which is differ from the parental type is called recombination. It is produced by crossing over. The frequency of recombination is higher for genes located apart on a chromosome than those located nearer (i.e. the recombination depends on the distance between the two genes on a chromosome).

Polygenic Inheritance:- A character whose expression is brought out by a no. of genes is called polygenic inheritance. The expression is due to cumulative effect of all the genes.

Eg. i) Kernel colour in wheat

ii) Human skin colour, height, eye colour .

In human, skin colour is control by three dominant gene A, B and C. So skin colour varies from dark in AABBCC to a very light in aabbcc individual. A

person with AaBbCc will have an intermediate colour. Thus the no. of alleles in the genotype would determine the darkness or lightness of the skin in an individual.

Pleiotropy:- It is the ability of a gene to have many effects or more than one phenotypic effect.

Eg. Sickled- cell –anaemia. The mutated gene produced abnormal haemoglobin and leads to sickled shaped RBC, reducing oxygen occurring capacity, tendency to clump blood etc.

SEX DETERMINATION

Autosome:-> Those chromosome which determine the somatic character of an individual is known as autosome.

Eg. Human has 46 chromosome i.e. 44+XY – in Male
44+XX – in Female
44= autosome,

Sex-chromosome:-> Those chromosome which determine the sex of an individual is known as sex-chromosome.

Eg. XX- female, XY-male.

The sex of an organism can be determined by the following ways.

i) Male heterogamy
ii) Female heterogamy
iii) Special type.

i) **Male heterogamy:->** Male individual produced two different types of gametes i.e. one posses X-chromosome and the other lacking X-chromosome while female will give similar gamete. It is of two types.
a) XX – XY Eg. Human, Drosophila
b) XX – XO Eg. Grass hopper, Bugs.

ii) **Female heterogamy:->** In this case, female will produced two types of gametes while male give similar gametes.

iii) Special type of sex determination :->

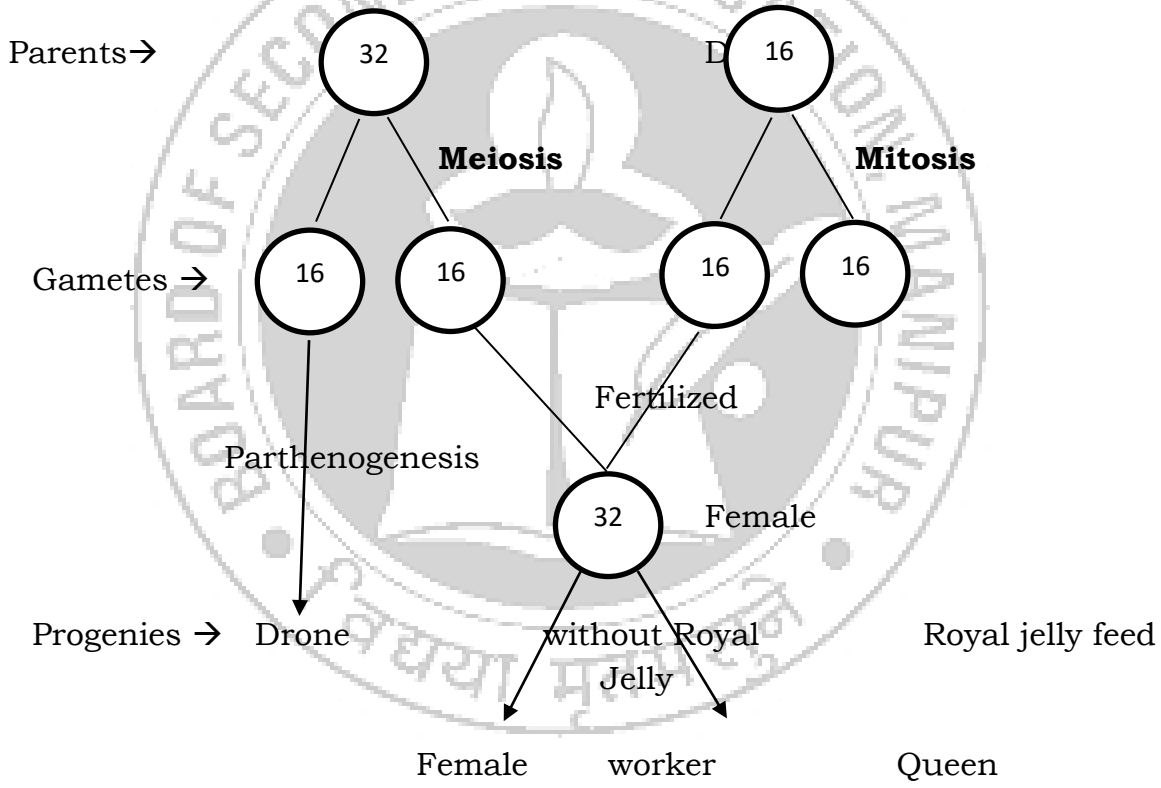


Fig: Sex determination in Honey bee

In this case the two sex chromosomes of female has been designated as Z and W chromosomes. In this organism females have one Z and one W where males have pair of Z chromosomes besides the autosomes. It is of two types

i) ZZ – ZW Eg. Birds, Reptiles.
ii) ZZ – ZO Eg. moth, butterfly.

iii) Special type (Male haploid, female diploid or hymenopteran type of sex determination)

In honey Bee, sex determination is based on the sets of chromosomes and individual receives. Three types of individuals are there in this colony.

- **i) Queen :-** They are fertile female and developed from fertilized egg i.e. (32= diploid chromosome).
- **ii) Worker :-** They are sterile female and developed from fertilized egg having 2 chromosome.
- **iii) Drone :-** They are male and developed from unfertilized egg by means of parthenogenesis having haploid (=16) chromosome. They produce sperm by mitosis.

i) Aneuploidy
- Hyperploidy – 2n+1= trisomy
  - 2n+2=tetrasomy
- Hypoploidy - 2n–1=Monosomy
  - 2n–2=Nullisomy

ii) Euploidy :-> In somatic chromosome, one or more additional haploid set of chromosome is present, then it is known as euploid.

Eg. Autotriploid – (AAA)
Autotetraploid- (AAAA)

MUTATION:-> It is a sudden and discontinuous changes that take place within the genome by which some parts of an organism changes to a greater or lesser extent.

Mutagen :-> Mutation causing agents are called mutagen.

Eg. UV-radiation, X-ray, suponate etc.

Mutation may be i) gene or point mutation.

ii) chromosomal mutation.

i) **Gene mutation:-** It is stable change in DNA. It may be change in a single base pair of DNA (muton).

Eg. Sickle-cell anaemia. Deletion and insertion of base pairs of DNA causes frame shift mutation.

ii) **Chromosomal mutation**:- It involves modification in the morphology. It alters the number and position of the existing gene. It is of two types.

a) **Morphological change** includes deletion, duplication insertion etc. of a segment of DNA causes morphological change.

b) **Numerical change:** It includes changes in the no. of existing chromosome i.e. chromosomal aberration. It may be i) Aneuploidy and ii) Euploidy.

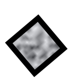
i) **Aneuploidy :** If chromosome no. increase by one or two or decrease by one or two, then it is called anaeuploidy.

Eg. 2n+1,2 or 2n-1,2

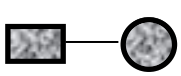
Pedigree Analysis

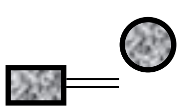
 - Male

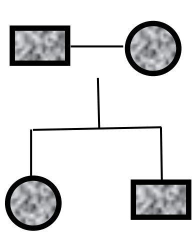
 - Female

 - Sex unspecified

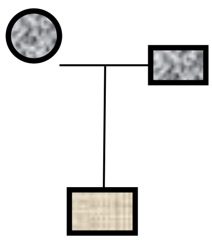
 - affected individual

 - mating

 - mating between relatives

 Parent

Children



Parent with
male child
affected with disease

FIG.: Symbols used in human pedigree analysis.

Example:

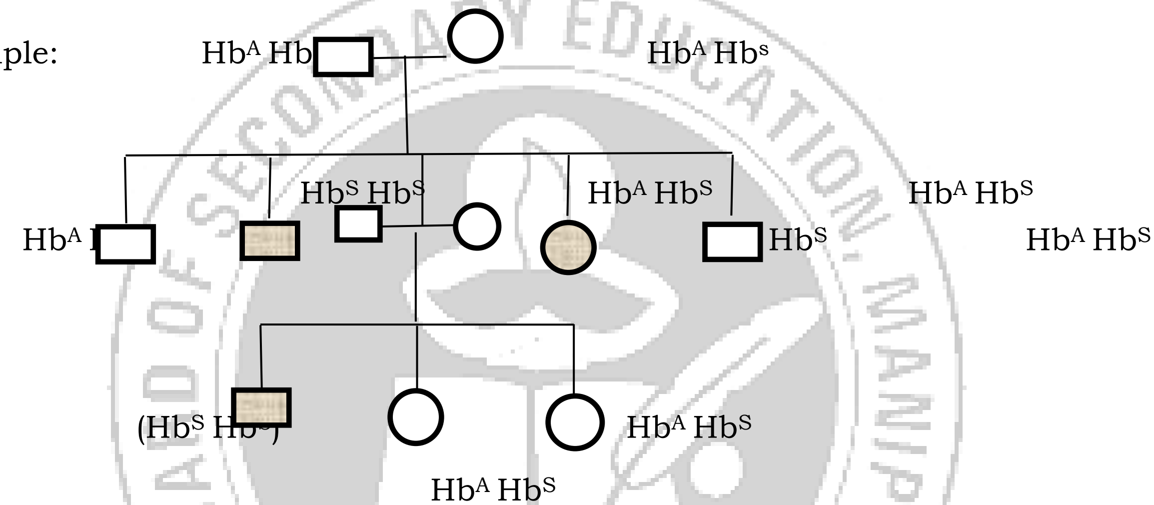


FIG.: Autosomal recessive trails (Sickle-cell anaemia)

Pedigree analysis :-> An analysis of traits in a several generations of a family is called pedigree analysis. The inheritance of a particular trait is represented in a family tree over generation. It provides a strong tools which help to trace the inheritance of specific trait, abnormality or disease.

GENETIC DISORDERS - i) Mendelian Disorder

ii) Chromosomal Disorder

Mendelian Disorder :-> It is caused by mutation in single gene. This disorder is transmitted to the offspring and pattern of inheritance can be traced in a family by pedigree analysis.

i) Colour blindness:- It is a sea-linked recessive disorder due to defect in red, green cone of eye resulting in failure to distinguish red and green colour.

The defected gene causes red, green colour blindness and they are present on X-chromosome.

A daughter may be colour blind only when mother is a carrier and father is a colour blind.

X^cX – mother carrier and X^cY – father colour blind.

ii) <u>Haemophilia:-</u> It is a sex-linked trait caused by recessive gene located at X-chromosome. These gene control the synthesis of a protein needed for clothing of blood. When the gene mutate, it loses its ability.

<u>Example- I</u>

X^hX^H (carrier female) X^HY (normal man)

X^h X^H X^H Y

X^hX^H	X^hY	X^HX^H	X^HY
Normal Daughter (carrier)	Haemophilic Son	Normal Daughter	Normal Son.

Gig. : If carrier woman marries a normal man, half of a son will be haemophilic and half are normal.

<u>Example- II</u> Colour blind mother Normal father

X^CX^C X Y

X^C X^C X Y

X^CX X^CY X^CX X^CY

Normal	Colour blind	Normal	Colour blind
Daughter	Son	Daughter	Son.

Gig. : Crossing between colour blind mother with normal father.

to produce the coagulant. Due to this, an affected individual for a simple cut will result non-stop bleeding. Heterozygous female (carrier) for haemophilia may transmit the disease to sons.

Queen Victoria was carrier of this disease.

SICKLED –CELL ANAEMIA: It is an autosomal linked recessive trait and it can be transmitted from parents to offspring when both the parents are carrier for gene (heteroxygous). This disease is caused at homozygous condition (Hb^SHb^S).

It is due to substitution of glutamic acid by valine of the haemoglobin molecule. So the shape of RBC disc shaped is changed into sickle shape thereby reducing oxygen carrying capacity.

Phenylketonuria: -> It is an autosomal recessive trait. The affected person lacks an enzyme phenylalanine hydroxylase needed to convert an amino acid phenylalanine to tyrosine. Accumulation of phenylalanine in brain causes mental retardation and hypopigmentation of skin.

Thalassemia: -> It is an autosome linked recessive blood disease transmitted from parents to offspring when both parents are carrier for the gene. It causes abnormal formation of haemoglobin result anaemia.

Chromosomal Disorder: →

i) **Down's Syndrome or Mongolsim- described by Langdon Down (1866):** It is trisomy of 21^{st} chromosome i.e. presence of an additional copy of the chromosome number 21. It is an autosomal disorder. (2n+1)=trisomy.

   The affected child is short statured with rounded face, prominent forehead, open mouth protending long tongue with short neck. Physical, psychomotor and mental development is also retarded.

ii) **Klinefelter Syndrome:→** It is a sex-chromosomal disorder due to the presence of an additional copy of X-chromosome resulting a Karyotype of 47 (XXY) = trisomy 2n+1 = 44 + XXY

   Symptom :->

   i) The individual has over all masculine development.

ii) Faminine development such as enlarged breast, faminine voice is also expressed.
iii) Under develop testis and body sparse hair.
iv) They are mentally retarded with sterile.

iii) Tuner's Syndrome:→ It is also sex-chromosomal disorders caused by the absence of one of the X-chromosome. i.e. 45 (XO + 44)

Symptom :->

(i) Such females are under develop overies, breast and small uterus.
(ii) Absence of menstrual cycle.
(iii) Mental retardation with abnormal intelligence.

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