Chapter-03 PRINCIPLES OF INHERITANCE AND VARIATION

Genetics

- It deals with the **inheritance,** as well as the **Variation of characters** from parents to offspring.
- It is the study of **genes and chromosomes**

Inheritance

- Inheritance is the process by which **characters are passed on from parent to progeny.** it is the basis of heredity
- Heredity is the tendency of offspring to resemble their parents

Variation

- It is the tendency of offspring to differ from their parents
- The main reason for variations are
  - **Crossing over**
  - **Mutation**
- Human knew from as early as **8000-1000BC** that one of the **cause of variation** was hidden in **sexual reproduction** .They exploited variation seen in nature (Plants and animal) to select organism with desirable characters. However, early humans had very little idea about the scientific basis of variation.

Eg: Well-known Indian breeds, **Sahiwal cow (Punjab)** **(**through artificial selection and domestication from ancestral wild cows)

GENETIC TERMS

- **Allele :**
  - They are alternative form of a gene
  - Genes which code for a pair of contrasting traits are known as alleles.
    Eg: T,t,R,r,Y,y
- **Phenotype**
  - The physical appearance of an organism is called Phenotype
  - The visible/observable characteristics of an organism is called phenotype
    Eg: Tall plant, blue eye, round seed

- **True breeding line**
  - A true breeding line is one that, having undergone continuous self-pollination, shows the stable trait inheritance and expression for several generation.
- **Genotype**

  The complete genetic constitution of anorganism is called Genotype.
  Eg: Tt,TT,RR,Rr,YY,Yy

- **Homozygous (True breeding/Pure line)**

  An organism with 2 identical allele of a gene
  Eg:TT,RR,YY,YY,rr,tt

- **Heterozygous**

  An organism with 2 different allele of a geneEg:Tt,Rr,Yy

Character and Trait

- A character is a heritable feature that variesamong individuals.
  Eg:Flower color,Plant Height,seed shape, Eyecolour.
- A trait is **a variant for character**,
  Eg: white or purple colors for flowers, Dwarfplant, Round seed, Blue eye.

Qn. An elephant always gives birth to a babyelephant and not some other animal, why ?
Ans:

Qn. A mango seed forms only a mango plant andnot any Other plant why ?
Ans:

GREGOR JOHANN MENDEL

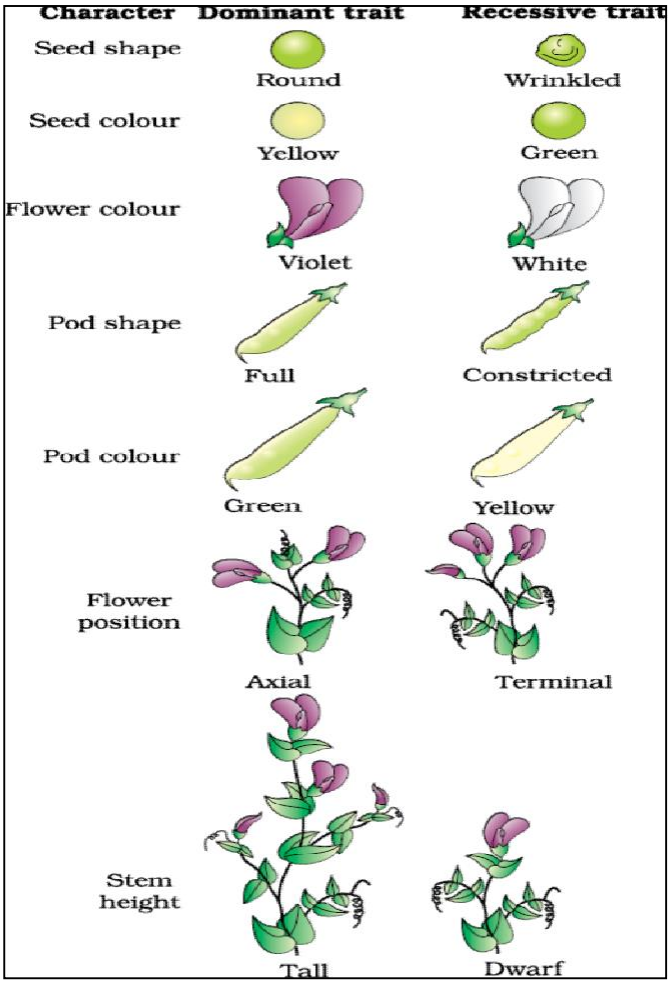
- He was an Austrian monk.
- He is known as father of genetics.
- He conducted **hybridisation experiment** on **garden pea plant (*Pisum sativum*)** for 7 years (**1856-1863**)
- Based on his experiment , he proposed **'laws of inheritance'** in living organisms .
- During Mendel's investigations into inheritance patterns , **it was for the first time**

that **statistical analysis and mathematical logic were applied to problems in biology**

- Mendel selected **14 true-breeding pea plant varieties**, as pairs which were similar except for **one character with contrasting traits.**
- The garden pea plant contains number of characters. Out of these, he selected and studied **only 7 characters**. Each of these 7 characters has 2 verities. The 7 characters are given below.

Sl No.	Character	Contrasting traits	
		Dominant	**Recessive**
1	**Height of the plants**	Tall	Dwarf
2	**Seed shape**	Round	Wrinkled
3	**Seed colour**	Yellow	Green
4	**Pod shape**	Inflated (full)	Constricted
5	**Pod colour**	Green	Yellow
6	**Flower position**	Axial	Terminal
7	**Flower color**	Violet	White

Contrasting traits studied by Mendel in Pea

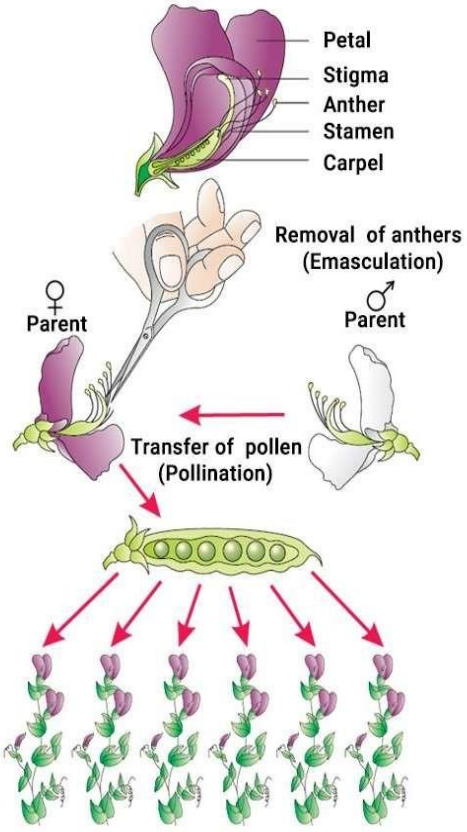


Reason for selecting Garden Pea plant

1. It has short life cycle so it gives quick results.
2. Plants shows clear contrasting character
3. Being a herb, it is Easy to cultivate
4. It has bisexual flower
5. It is generally self-pollinated and so self fertilised. However, it can be Cross pollination is easy if self-pollination is prevented.

Monohybrid cross- Inheritance of one gene

- It is the cross involving **two forms of a single character.**
- It is the simplest cross performed by Mendel
- Mendel conducted artificial pollination/cross pollination experiments using **several true-breeding pea lines.**
- Mendel crossed tall plants and dwarf pea plant to study the inheritance of one gene.
- He collected seed produced as a result of above cross and grew them to generate F1 (Filial 1 Progeny).

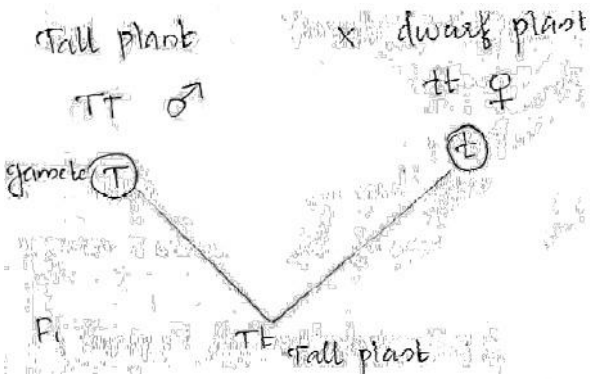


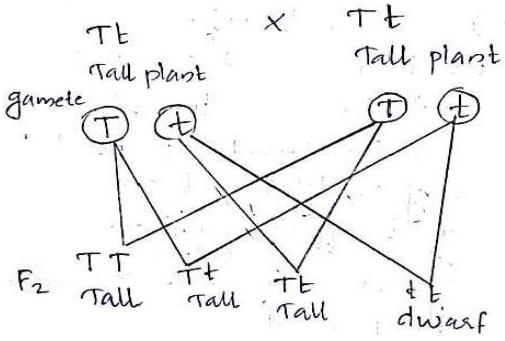
- Mendel observed that all the F1 progenies were tall (Like one of its parent and none were dwarf)

> He made similar observations for the other pairs of traits – he found that the F_1 always resembled either one of the parents, and that the trait of the other parent was not seen in them.

- He then self pollinated the F1 progeny (Tall plants) to generate F2. He observed that **75% of the F2 progenies were tall and 25% were dwarf (Ie: 3:1)**
  Ie: characters that was not seen in the F1 generation expressed in the F2 (dwarf)
- He also found that ,The tall and dwarf traits were **identical to their parental type** and **did not show any blending**, that is all the offspring were either tall or dwarf, none were of in between height, No blending of characters in offsprings.
- Based on this observation ,Mendel proposed that something being was being stably passed down unchanged from parents to offspring through gametes over successive generation. Mendel called it as **factors.** Now we called them as genes.
- Genes, therefore, **are the units of inheritance. They contain the information that is required to express a particular trait in an organism.** Genes is a chemically a segment of DNA (RNA in some virus .

Example-1

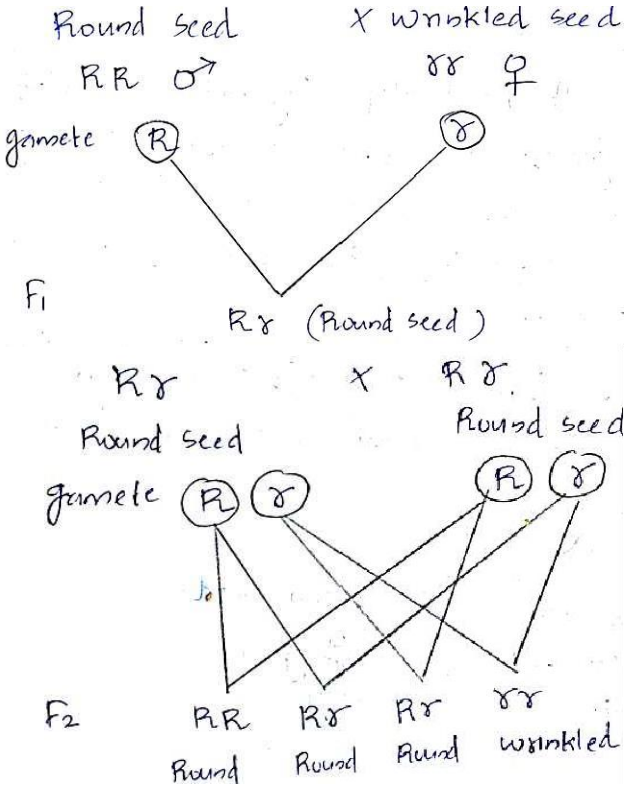




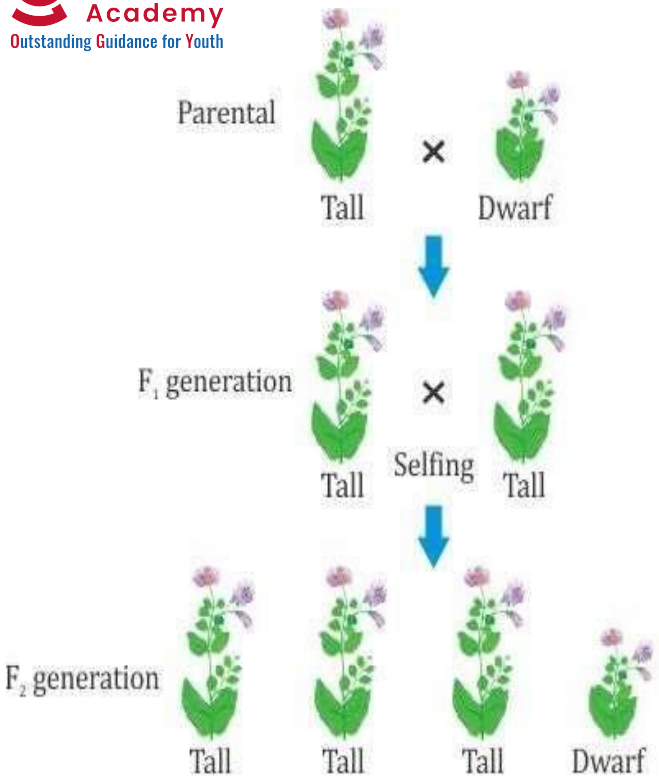
Monohybrid genotypic ratio= 1:2:1
Monohybrid phenotypic ratio =3:1

Similar results were obtained with the other traits that he studied (Seed shape,flower colour etc.): **only one of the parental traits was expressed** in the F1 generation while at the F2 stage both the traits were expressed in the proportion 3:1. **The contrasting traits did not show any blending at either F1 or F2 stage**

Example-2



Monohybrid genotypic ratio= 1:2:1
Monohybrid phenotypic ratio =3:1

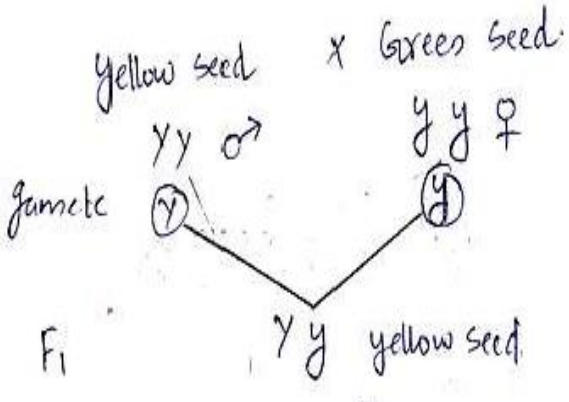


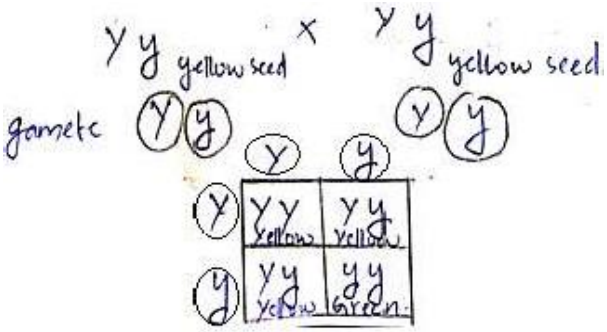
Diagrammatic representation of monohybrid cross

PUNNET SQUARE

- It was developed by **British geneticist Reginald C Punnet**
- **It is the graphical representation to calculate the probability of all possible genotype of an offspring in a genetic cross .**
- The possible gametes are written on 2 sides, usually on the top row and left column. All possible combinations are written in boxes below in square, which generates a square output form

Example



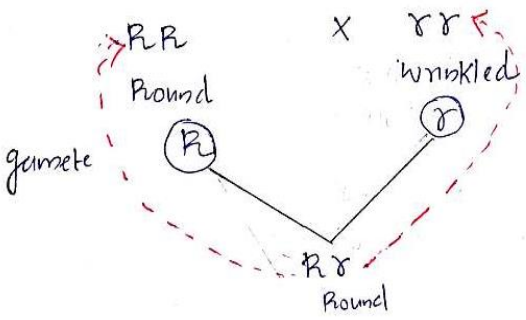


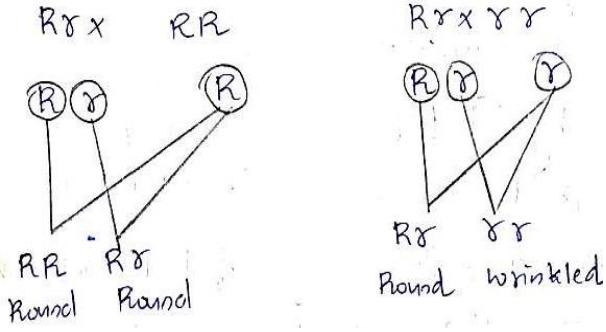
Monohybrid genotypic ratio= 1:2:1
Monohybrid phenotypic ratio =3:1

BACK CROSS & TEST CROSS

Back cross

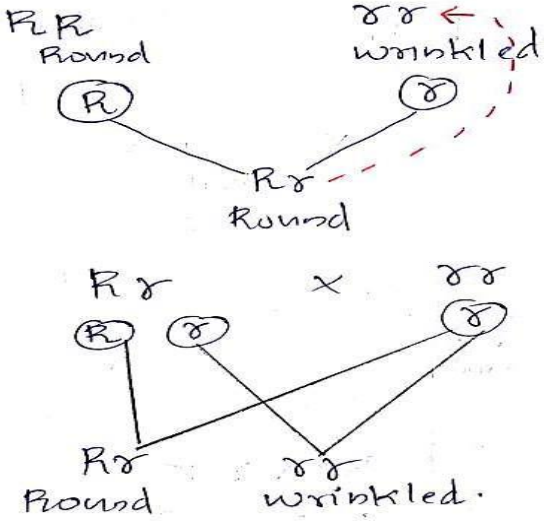
- It is the cross of F1 progeny with **one of its** parent





Test Cross

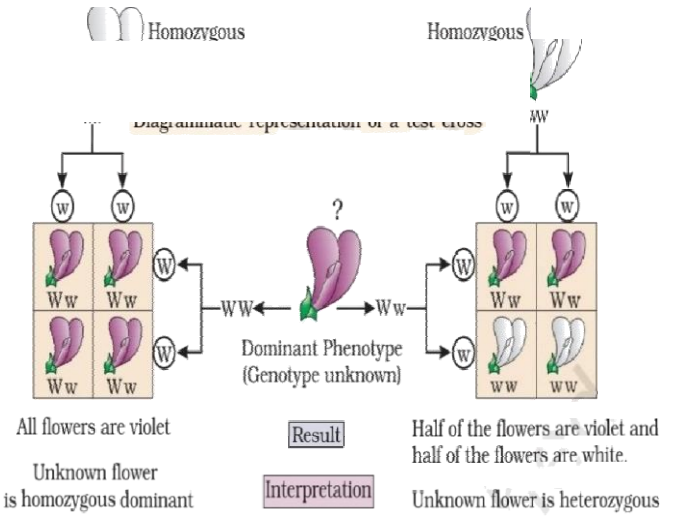
- It is the crossing of (F1) progeny with its **recessive parent**.
- It is used to **find unknown genotype** of an individual.



Monohybrid Test cross ratio= 1:1
Dihybrid test cross ratio=1:1:1:1

Qn. What will be the genotype of a tall plant ,If it produce tall plant and dwarf plant in the ratio 1:1 during test cross.?

Ans:



Diagrammatic representation of a test crosses

Mendel's Laws on Inheritance (Principles of inheritance)

Based on **his observations on monohybrid crosses**, Mendel proposed **two general rules** to consolidate his understanding of inheritance in monohybrid crosses. **Today these rules are called the Principles or Laws of Inheritance:**

1- The First Law or Law of Dominance
2- Second Law or L w of Segregation

1. Law Of Dominance (1st law)

The main points are ...

I. **The characters are controlled by discrete units called factors.**
II. **Factors occur in pair.**
III. **In a dissimilar pairs of factors (Heterozygous) ,one member of pairs dominates over the other. (The dominated one is called Dominant , and other character is called Recessive)**

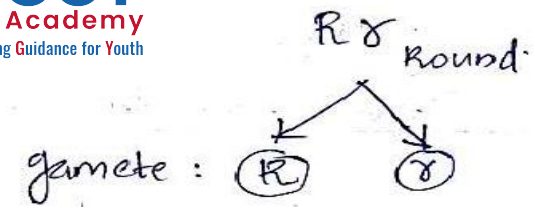
- This law is used to explain the expression of only one of the parental character in the F1 of monohybrid cross and expression of both in F2
- This law explains the proportion of 3:1 obtained at the F2

2. Law of segregation

(2nd law/law of purity of gamete)

- This law is based on the fact that the **alleles do not show any blending** and that both the characters are **recovered** as such in the **F2 generation** though one of these is not seen at the F1 stage.

- This law states that,
  "During gamete formation 2 factors for a trait present in an individual will separate from each other and enter into each gamete"

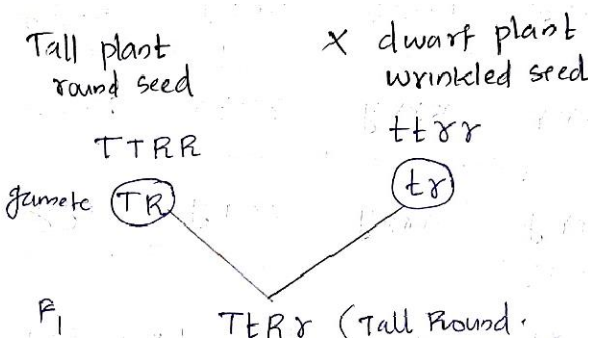


- Thus, a homozygous parent produces all gametes that are similar.
- while a Heterozygous one produces two kinds of gametes each having one allele with equal proportion.

- Both 1st and 2nd law of Mendel obtained from monohybrid cross.

Dihybrid phenotypic ratio=9:3:3:1
Dihybrid genotypic ratio = 1:2:1:2:4:2:1:2:1

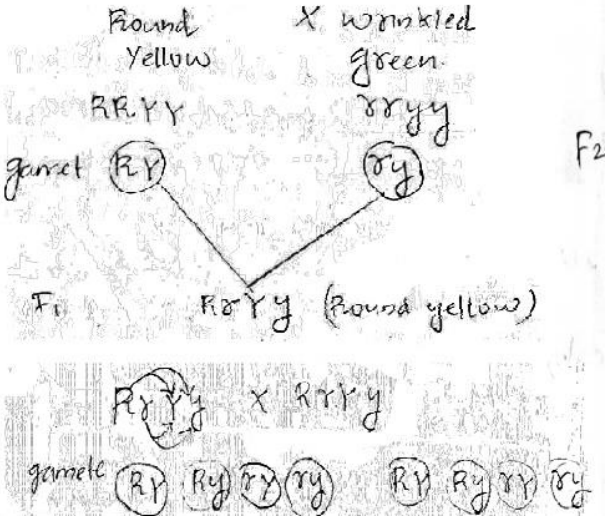
Example-2

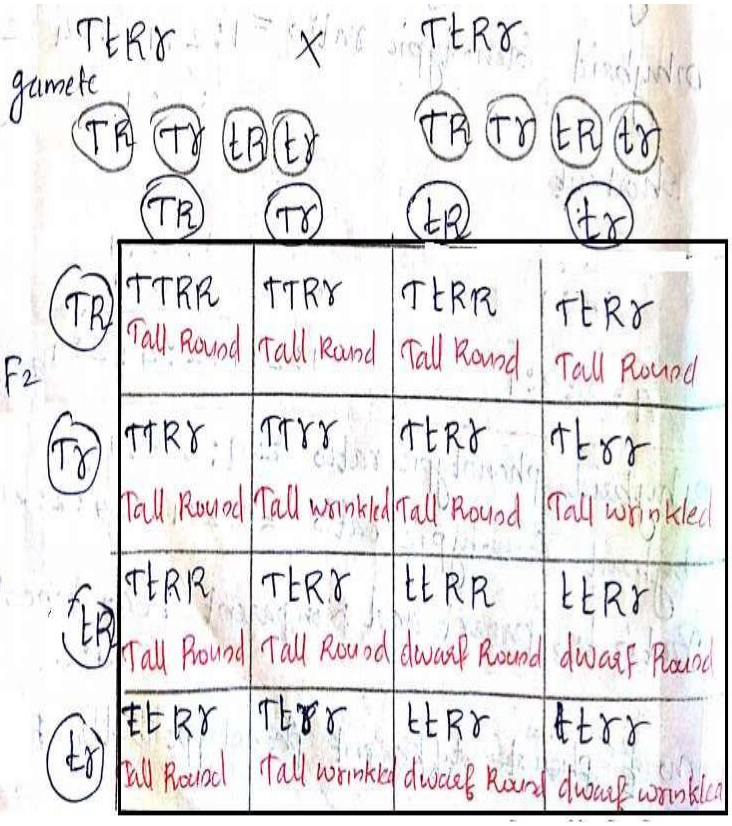


Dihybrid cross-Inheritance of 2 genes

"It is a cross involving **2 characters**/a cross involving plants differing in 2 characters"

Example-1





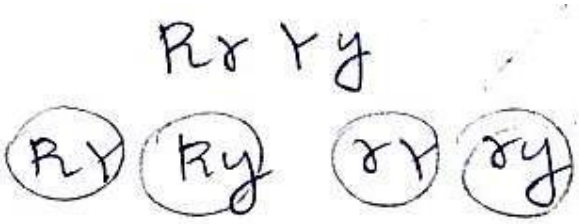
- **Dihybrid phenotypic ratio=9:3:3:1**
- **Dihybrid genotypic ratio =1:2:1:2:4:2:1:2:1**
- **Ratio b/w parent and nonparent (recombinant) =10:6**
- **Number of different phenotype in the F2 dihybrid cross = 4**
- **Number of different Genotype in the F2 dihybrid cross = 9**

Qn. Write the different gametes produced from the plant with genotype TtRr ?

Ans:

3. <u>Law of Independent assortment</u>

- This law is formulated from Dihybrid cross
- This law states that
- "'when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters"
- This means that the inheritance of one trait is not dependent on the inheritance of another trait



- **This law is not applicable for the genes located on the same chromosome**
  Ie: Linked gene.
- **Linked genes are exception to mendelian principle**

DEVIATION FROM MENDLIAN PRINCIPLE / NON MENDELIAN INHERITANCE

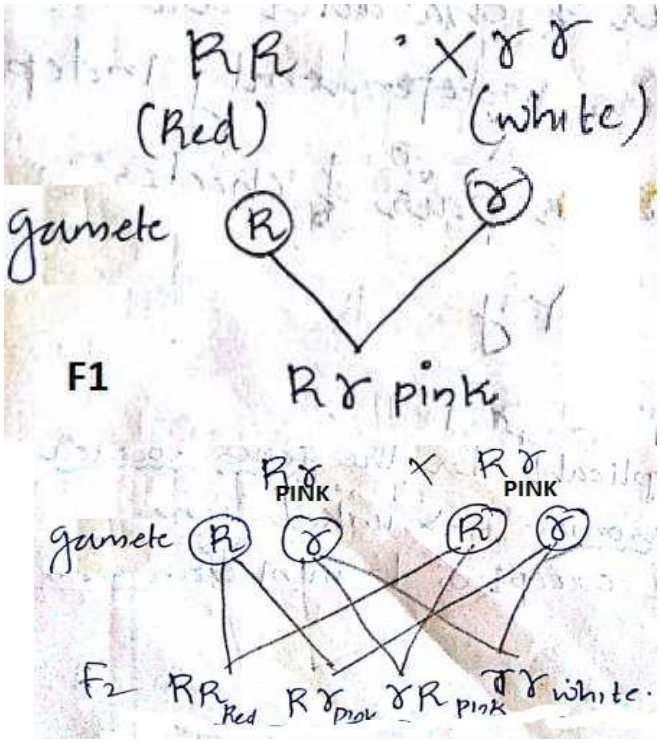
- Non-Mendelian inheritance is any pattern of inheritance in which traits **do not segregate in accordance with Mendel's laws**

1. Incomplete dominance

- It is the inheritance in which the heterozygous offspring show **intermediate character between 2 parents.**

Example-1

- Inheritance of flower colour **in the dog flower (snapdragon or *Antirrhinum sp.)*** and *Mirabilis jalapa* (4 o' clock plant- not mentioned in text book) is a good example to understand incomplete dominance. It was studied by Carl Correns of Germany .

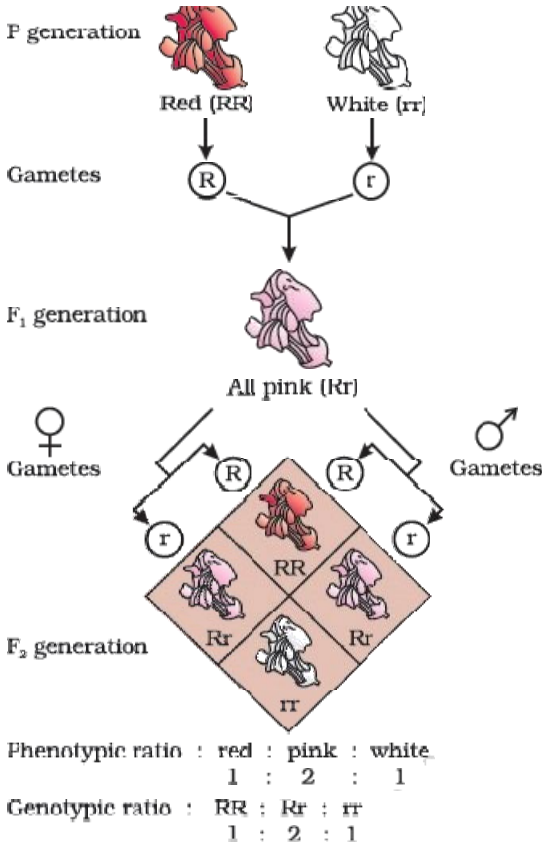


Phenotypic ratio=1:2:1
Genotypic ratio=1:2:1

- When crossed a true-breeding red-flowered (RR) *Antirrhinum* and true breeding white-flowered *Antirrhinum* plants (rr), the F1 (Rr) was **pink**
- When the F1 was self-pollinated, **the F2** resulted in the following ratio

1(RR) Red: 2 (Rr) Pink: 1 (rr) White.

- Here the genotype ratios were exactly as **we would expect in any mendelian monohybrid cross, but the phenotype ratios had changed from the 3:1 dominant: recessive ratio.**
- What happened was **that R was not completely dominant over r and this made it possible to distinguish Rr as pink** from RR (red) and rr (white) .**This is due to incomplete dominance. So the heterozygous offsprings** shows intermediate character (Pink) **between 2 parents.**



Example-2

Starch grain size in Pea seed

- Starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b)
- BB ----- →starch synthesized effectively (Large sized starch grains)

- bb ------- → lesser efficiency in starch synthesis (Small sized starch grains)
- **Bb------→ Intermediate sized starch grains**

2. Co Dominance

- Here **both alleles of gene are expressed** in heterozygous condition.
- In the case of co-dominance, the F1 generation **resembles both parents**

Example-1

ABO Blood group

- ABO blood group is controlled by the gene 'I'
- 'I' gene has 3 alleles –I^A ,I^B, i
- The allele I^A and,I^B produce slightly different form of the sugar .
- This sugar are protrudes from the plasma membrane of RBC.
- 'i' donot produce sugar
- When '$I^{A'}$ and 'i' are present in an organism (I^Ai), only I^A expressed because 'i' donot produce any sugar .
- When '$I^{B'}$ and 'i' are present in an organism (I^Bi), only I^B expressed because 'i' donot produce any sugar .
- **When I^A and I^B are present in an organism (I^AI^B), they both express their own type of sugars. This is due to co dominance. Such RBC contains both sugar 'A' and 'B' type of sugars.**

Blood Group (Phenoype)	Genotype
A	I^AI^A,I^Ai
B	I^BI^B,I^Bi
AB	**I^AI^B**
O	ii

- There are **4 different phenotype** present in ABO blood group
- There are **6 different genotype** present in ABO blood group

Qn. Find out the genotype of children/s born to Parents with blood group 'AB 'and 'O' ?
Ans:

3. Multiple alleles

- Some genes have more than 2 alleles. This phenomenon is called multiple allelism .
- Here we can see that there are more than two alleles (I^A ,I^B, I), governing the same character (Blood group)

Example

ABO Blood group

- ABO blood group is controlled by the gene 'I'
- 'I' gene has 3 alleles –I^A ,I^B, i
- These alleles are located at the same locus in a given pair of homologus chromosome .

4. Pleiotropy

- Multiple effect of a gene is called **pleiotropy. Such genes are called Pleiotropic gene**
- **Here single gene may produce more than one effect.**
- The underlying mechanism of Pleiotropy in most case is the effect of a gene on metabolic pathway-which produce different phenotype.

Example-1

- Starch synthesis in pea seeds
- It has two alleles (B and b).

Genotype	Phenotype	
	Starch grain size	**Seed shape**
BB	Large	Round
bb	Small	Wrinkled
Bb	Intermediate	Round

- Here a single gene control both starch grain size and seed shape

Example-3

PKU (Phenykenonuria)

- The disease is caused by the mutation in the gene that code for the enzyme **phenylalanine hydroxylase** (Single gene mutation)
- It result mental retardation, reduction in **hair, pigmentation in skin** in patients.

5. Polygenic inheritance

- **A character** whose expression is controlled by **number of genes** is called polygenic inheritance .
- Besides the involvement of multiple genes, polygenic inheritance also takes into account the **influence of environment.**
- In a polygenic trait the phenotype reflects the contribution of each allele, i.e., **the effect of each allele is additive**

Example-1

Human Height

Example-2

Human Skin Colour

- Skin color is controlled by 3 pairs of gene (A,B,C)
- AABBCC--→Darkest Skin color
- AaBbCc--→Intermediate skin color
- Aabbcc--→Lightest skin color
- **The number of each type of alleles in the genotype would determine the darkness or lightness of the skin in an individual.**

Publication Of Mendel's Work

- Mendel published his work his work in **1865.**
- But scientists of his time did not accept his theory because

Reason for non-acceptance of Mendel's work

i. Communication was not easy (as it is now) in those days and his work could not be widely publicised.
ii. He used maths to explain biological phenomenon was totally new and unacceptable to many of the biologists of his time
iii. He could not provide any physical proof for the existence of factors or what they were made of.
iv. His concept of genes (or factors, in Mendel's words) as stable and discrete units that controlled the expression of traits and, of the pair of alleles which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature..

Reason for Mendel's success

1. Gradual planning
2. Attention was focused only on one character at a time
3. Maintenance of accurate record of result obtained
4. Careful experimentation and observation
5. His experiments had a large sampling size, which gave greater credibility to the data that he collected
6. He was a lucky person (didn't find linkage phenomenon

Rediscovery of Mendelian Principle

- In 1900, three scientist namely
  - **Carl correns**
  - **Hugo De Vries**
  - **Von Tschermak**

independently rediscovered mendel's results on the inheritance of characters.

- By this time microscope are advanced.
- Scientist observed cell division.
- This lead to the discovery of a structure in the nucleus that appeared to double and divide **just before cell division**, these were called **chromosome (Colored body- because that can be visualised only by staining)**
- **By 1902** chromosomal movement during meiosis had been worked out.

Chromosomal theory of Inheritance

- Proposed by **Walter Sutton and Theodore Bovery** In 1902
- Walter Sutton and Theodore Boveri noted that the **behavior of chromosomes was parallel to the behavior of genes.** (The important things to remember are that chromosomes as well as genes occur in pairs. The two alleles of a gene pair are located on homologous sites on homologous chromosomes)
- They used chromosome movement to explain Mendel's law.
- They studied behavior of chromosome during mitosis and meiosis.
- Paring and separation of a pair of chromosome will lead to segregation of a pair of factor they carried"
- Sutton united chromosomal segregation with Mendelian principles and called it as **chromosomal theory of inheritance.** It states that "Genes are located on

chromosomes and they later segregate and independently assort during meiosis"

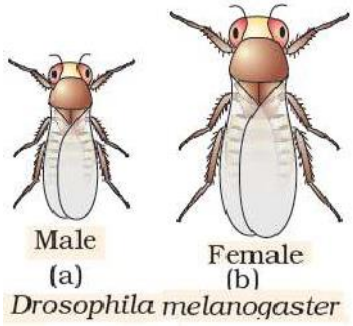
- The chromosomal theory of inheritance provided a physical explanation for Mendel's laws of inheritance and revolutionized our understanding of genetics

Experimental verification of chromosomal theory of inheritance

- The experimental verification of the chromosomal theory of inheritance by **Thomas Hunt Morgan and his colleagues, led to discovering the basis for the variation** that sexual reproduction produced
- He conducted his experiment on **tiny Fruit fly (*Drossophila melanogaster*)**

Reason for selecting fruit fly

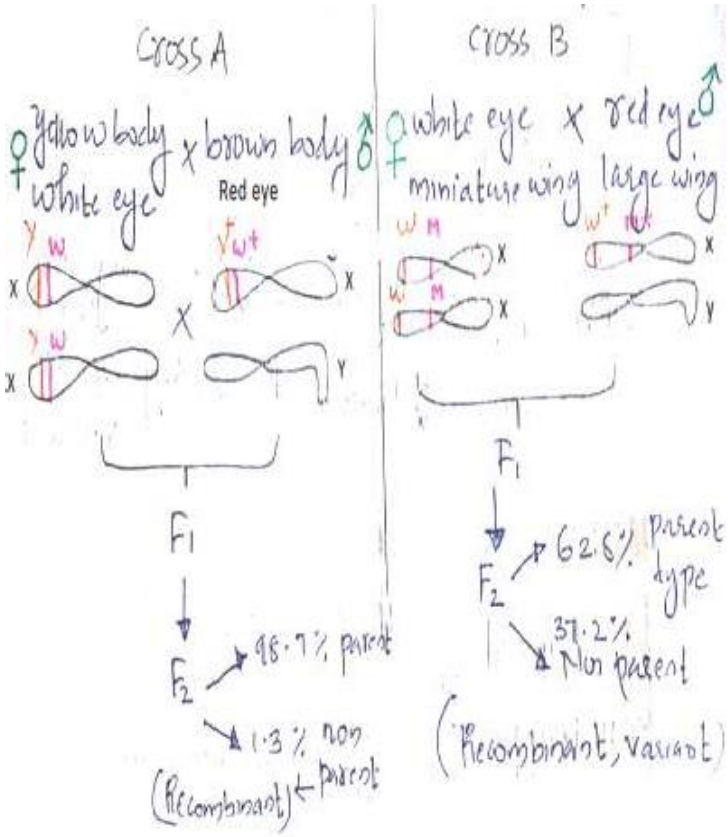
i. It can grown on simple synthetic medium in the laboratory
ii. They complete their life cycle in about two weeks,
iii. A single mating produce large number of progeny flies
iv. There is clear difference between male and female. **(females are larger than male).**
v. It has many types of Hereditary variations, that can be seen with low power microscopes.



Drosophila melanogaster

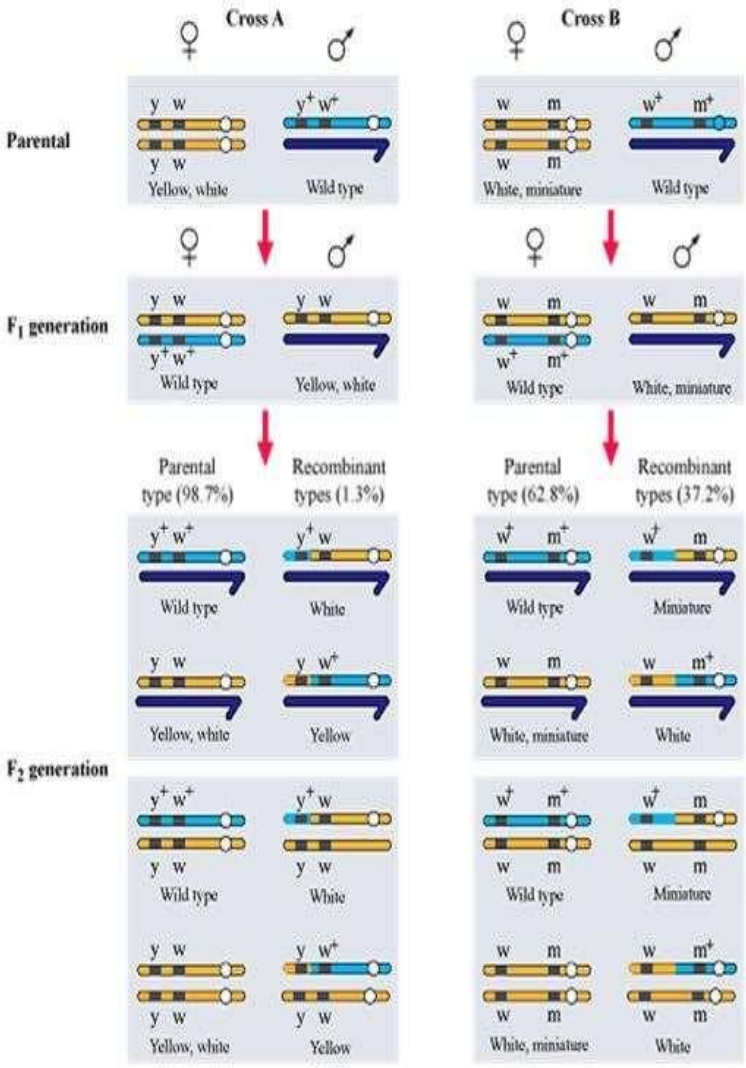
6. LINKAGE

- Morgan carried out several **dihybrid crosses in Drosophila** to study genes that were sex-linked. These crosses were similar to the dihybrid crosses carried out by Mendel in peas.
- Morgan hybridised **yellow-bodied, white-eyed females to brown-bodied, red-eyed males (Wild type)** and intercrossed their F1 progeny. He observed that the **two genes did not segregate independently** of each other and the F2 ratio **deviated** very significantly from the **9:3:3:1 ratio** (expected when the two genes are independent).
- Morgan and his group knew that the genes were located on the **X chromosome** and saw quickly that **when the two genes in a dihybrid cross were situated on the same chromosome, the proportionof parental gene combinations were much higher than the non-parentaltype.**



- In the cross A , F2 ratio is deviated from normal Mendelian dihybrid ratio (10:6). It is due to linkage.

- **The physical association of genes in a chromosome is called linkage, such genes are called Linked genes .**
- **The term linkage and recombination coined by. T H Morgan.**
- **Linked genes are exception to law of independent assortment .**
- Morgan found that the genes white and yellow were very tightly linked and showed only 1.3 per cent recombination while white and miniature wing showed 37.2 per cent recombination
- Ie: In the above cross, the genes for yellow body, white eye and gene for brown body ,red eye are located on X chromosome (Sex linked/X linked genes) **and are tightly linked genes**
- **Tightly linked genes shows low recombination (Non parent type)**
- **Loosely linked genes show high recombination**



Mapping of genes

- Proposed **by Alfred Sturtevant (Student of T.H Morgan)**
- He mapped position of genes in a chromosome
- He used frequency of recombination between gene pairs on the same chromosome as a measure of distance between genes.
- Today genetic maps are used as a starting point in the sequencing of whole genomes as was done in the case of the Human Genome Sequencing Project (HGP)

Sex Determination

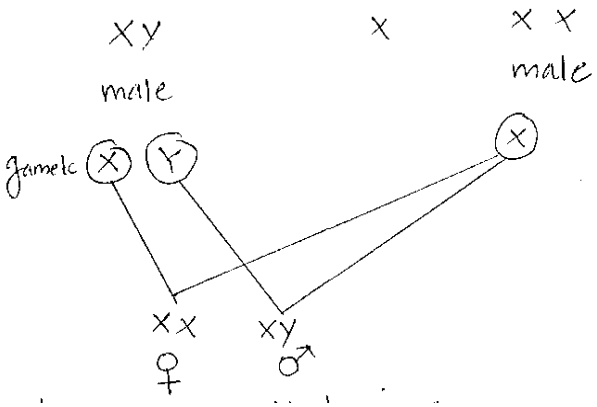
- The chromosome involved in the sex determination is called **sex chromosome** (Allosome). It include 'X' and 'Y' chromosome.
- Autosomes are chromosome present in an organism other than sex chromosome.
- The number of autosomes are same in both male and female of same species.
- The initial clue about the genetic/chromosomal mechanism of sex determination can be traced back to some of the experiments carried out in **insects**
- **Henking (1891)** studied spermatogenesis **in some insects**.
- He observed that 50% of sperm received a nuclear structure after spermatogenesis, other 50% of sperm did not received it .
- Henking called these nuclear structure **as 'X body'**, (now it is called as X-chromosome). but he could not explain its significance

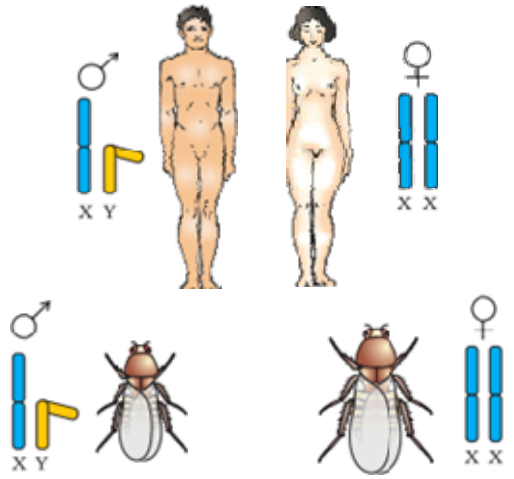
Mechanism of sex determination

- Various types of sex determinations are given below
  a) XX-XY mechanism–Human being, drosophila
  b) ZZ-ZW mechanism-Birds
  c) XX-XO mechanism-Insects
  d) Haplo-Diplody mechanism-Honey bee

a)XX-XY mechanism –Human being, drosophila

- Here both male and female have same number of chromosomes.
- Males (XY) and produce 2 types of gametes (Sperms-**Heterogametic**) containing 'X' Or 'Y' chromosomes , besides Autosomes.
- Females are homozygoys (XX) and produce only one type of gamete (egg/ovum) containing only 'X' chromosomes, besides Autosomes.
- Sex of the baby is determined by the type of Sperm entering into the egg
- If the sperm containing 'X' chromosome enter into egg female baby is produced
- If the sperm containing 'Y' chromosome enter into egg, male baby is produced.
- So sex of the baby is determined by the **father** not the mother in XX-XY mechanism.

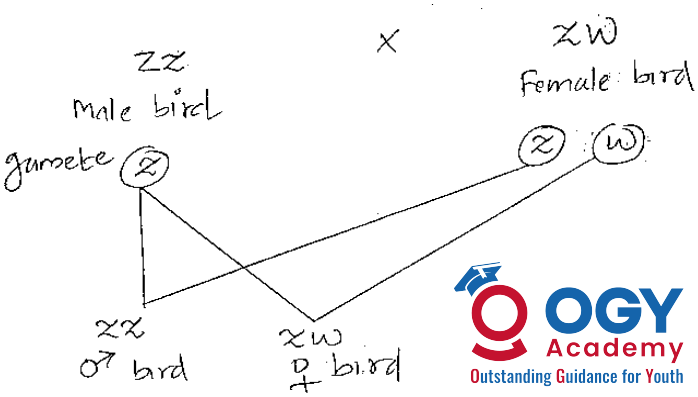


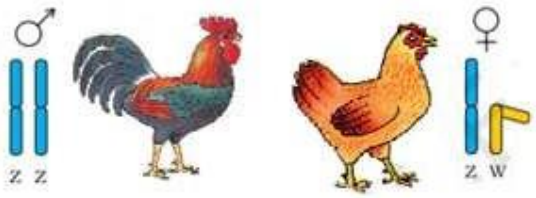


b) ZZ-ZW mechanism-Birds

- Here both male and female have same number of chromosomes.
- Females (ZW) produce 2 types of gametes (Eggs-**Heterogametic**) containing 'Z' or 'W' chromosomes besides Autosomes.
- Males (ZZ) produce only one type of gamete (Sperms) containing only 'Z' chromosomes besides Autosomes.
- Sex of the baby is determined by the type of Egg into which sperm enter
- If the Egg containing 'Z' chromosome Receives a sperm, male baby is produced
- If the Egg containing 'W' chromosome Receives a sperm, Female baby is produced

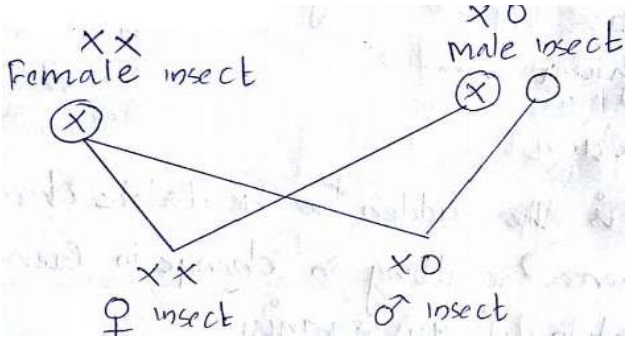
- So sex of the baby is determined by the **Mother** not the Father in ZZ-ZW mechanism.

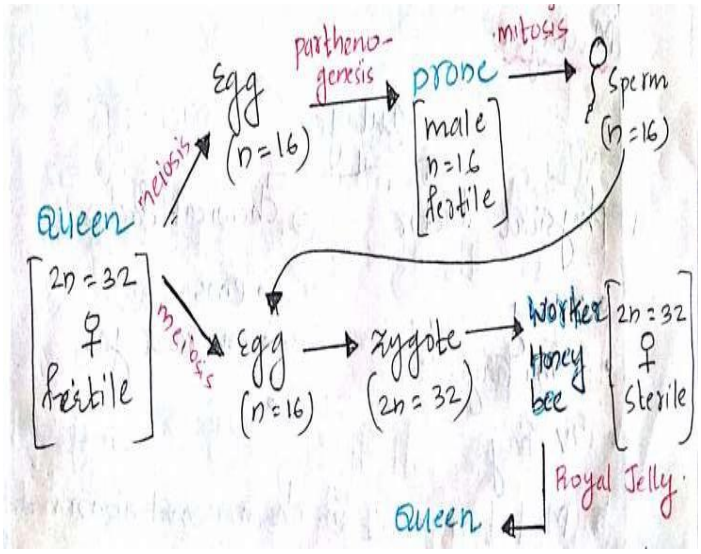




c) XX-XO mechanism-Insects (Grasshopper)

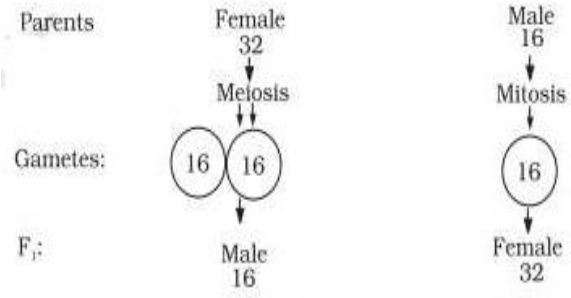
- Here males are one chromosome less than (Sex chromosome)that of females
- Females are homozygous (XX) and produce only one type of gamete (Egg/Ovum) containing only 'X' chromosome besides autosomes.
- males (XO) produce two types of gametes (Sperms).50% of sperms contains 'X' chromosomes besides autosomes, the other 50% sperms contains only autosomes (Sex chromosome absent)
- So sex of the insect is determined by the type of sperm enter in to the egg.
  Ie: **Male insect** will determine the sex of the baby





- The sex determination in honey bee is based on the number of sets of chromosomes an individual receives
- Here an offspring formed from the union of a sperm and an egg develops as a female (queen or worker), and an unfertilized egg develops as a male (drone) by means of parthenogenesis.
- Here **females are diploid** (32 chromosomes) and **males are Haploids** (16 chromosomes), this type of sex determination is called haplo-diploidy sex determination

- Here males (Drone) produce sperms by mitosis. They do not have father and thus cannot have sons, but have grandfather and have grandsons.



Sex determination in honey bee

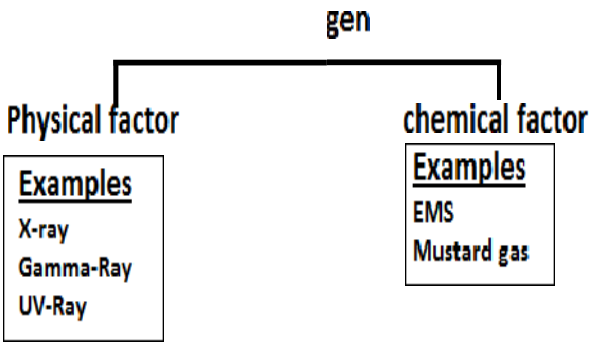
Qn. in our society women are blamed for giving birth to female children , Evaluate this statement
Ans: Humans have 23 pairs of chromosomes, with one pair determining sex (XX for females, XY for males).The sperm contributes the deciding factor. Sperm cells carry either an X or a Y chromosome. When a sperm fertilizes an egg (which always carries an X chromosome), the outcome determines the baby's sex:

- **X sperm + X egg (XX):** This results in a female child.
- **Y sperm + X egg (XY):** This results in a male child.

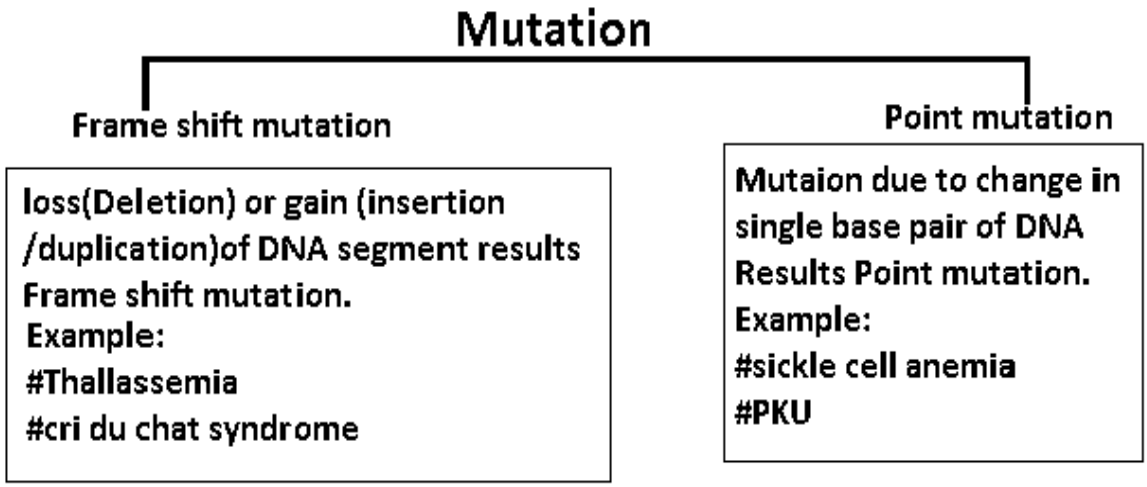
Therefore, the father's sperm determines the baby's sex, not the mother's egg. Blaming women for the child's gender is not only scientifically inaccurate.

MUTATION

- Mutation is a phenomenon which results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism.
- **In addition to recombination, mutation is another phenomenon** that leads to **variation in DNA.**
- The substance that cause mutation is called Mutagen

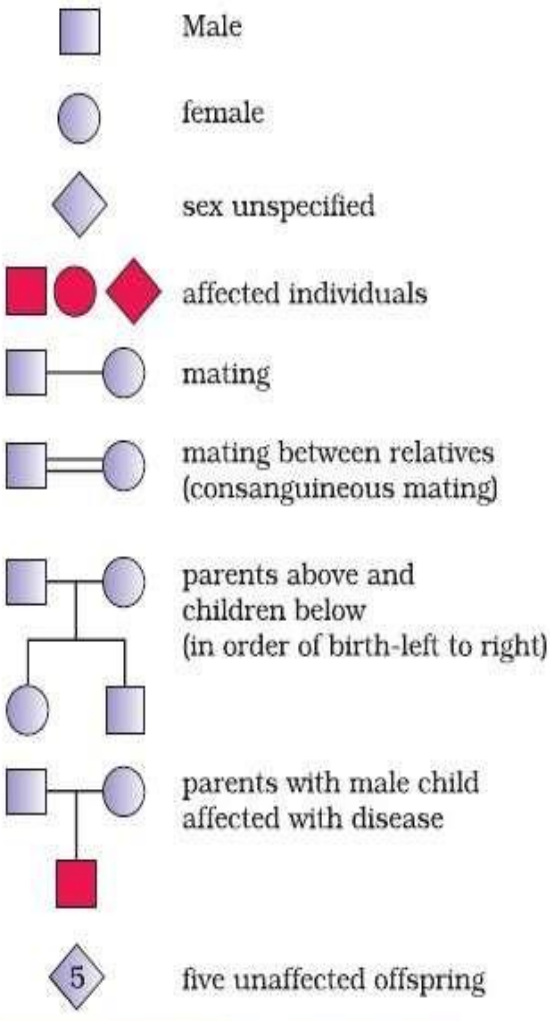


- Loss (deletions) or gain (insertion/duplication) of a segment of DNA, result in alteration in chromosomes. Since **genes are known to be located on chromosomes**, alteration in chromosomes results in abnormalities or aberrations **(Chromosomal aberrations**).
- Chromosomal aberrations are seen in **cancer cells**.
- Mutations are of 2 types
  a) Point mutation
  b) Frame shift mutation



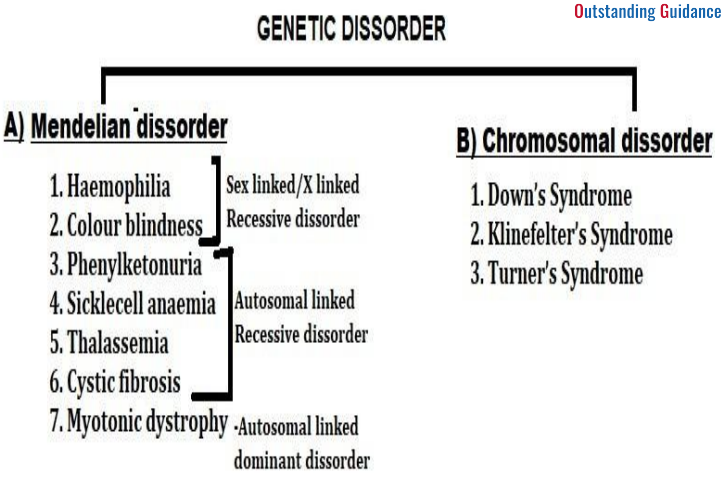
PEDIGREE ANALYSIS

- It is the analysis of trait in a several generations of a family is called pedigree analysis.
- Here inheritance of a particular trait is represented in the family tree (Chart showing family history) over generation.
- This analysis is used to trace the inheritance of **a specific trait or abnormality or a disease**
- In human genetics, pedigree study provides a strong tool, **which is utilised to trace the inheritance of a specific trait, abnormality or disease**
- The symbols used in pedigree analysis is given below



Symbols used in the human pedigree analysis

GENETIC DISSORDERS



A) Mendelian dissorder

- It is due to **mutation or alteration** in the single gene.
- This disorder are **transmitted** to the offspring as we studied **in the principles of inheritance**
- This disorder can be traced in a family using **pedigree analysis.**
- Mendelian disorders may be dominant or recessive. By pedigree analysis one can easily understand whether the trait is dominant or recessive

1. HAEMOPHILIA/BLEEDER'S DISEASE/ROYAL DISEASE

- It is a **sex linked (X-linked) recessive** disease
- Here a single protein that is a part of chain (cascade) of protein involved **in clotting of blood is affected**. Due to this, in affected individual a simple cut will result nonstop bleeding

Genotypes are

- Normal male X^HY
- Normal female X^HX^H
- Hemophilic male X^hY
- Hemophilic female X^hX^h
- Hemophilic carrier (Female-) X^HX^h

- The disease is transmitted from an unaffected carrier female (X^HX^h)to some of male progeny
- The possibility of a female becoming hemophilic is extremely rare, because mother of such female has to be at least carrier and father should be hemophilic (He is unviable in the later stage of life)
- **The family pedigree of Queen Victoria shows number of hemophilic descends. she was a carrier for this disease**.

2. COLOUR BLINDNESS

- It is **a sex linked (X-linked)recessive** disease
- It is due to defect in **either red or green cone of eye** resulting **in failure to discriminate between red and green colour**
- This defect is due to mutation in certain gene present in the X- Chromosome

The genotypes are

- Normal male X^CY
- Normal female X^CX^C
- Color blind male X^cY
- Color blind female X^cX^c
- Color blind carrier (Female) X^CX^c

- It occurs 8% of male and 0.4% of female
- This is due to gene mutation for red and green colour. They are located on X-Chromosome. Males have only one X-Chromosome and female s have two X-chromosome.
- The son of a woman who carries the gene has a 50 % chance of being colour blind.
- A daughter will not be normally colour blind unless her mother is a carrier and father is a color blind.
- **X-linked recessive trait shows transmission from carrier female to male progeny.**

3. PKU (PHENYLKETONURIA)

- This is **the autosomal linked recessive trait.**
- PKU is an **inborn error in amino acid metabolism**
- The affected individual **lacks an enzyme (phenylalanine hydroxylase)** that converts the amino acid **phenylalanine into tyrosine**.
- As a result of this phenylalanine is accumulated and converted into **phenylpyruvic acid** and **other derivatives**.
- Accumulation of these in brain results in mental retardation. These are also excreted through urine because of its poor absorption by kidney.

The genotypes are

- Normal AA
- Carrier Aa
- Affected aa

- This disease is transmitted from parents to the offspring when both parents are carried (Heterozygous)

4. SICKLE CELL ANAEMIA

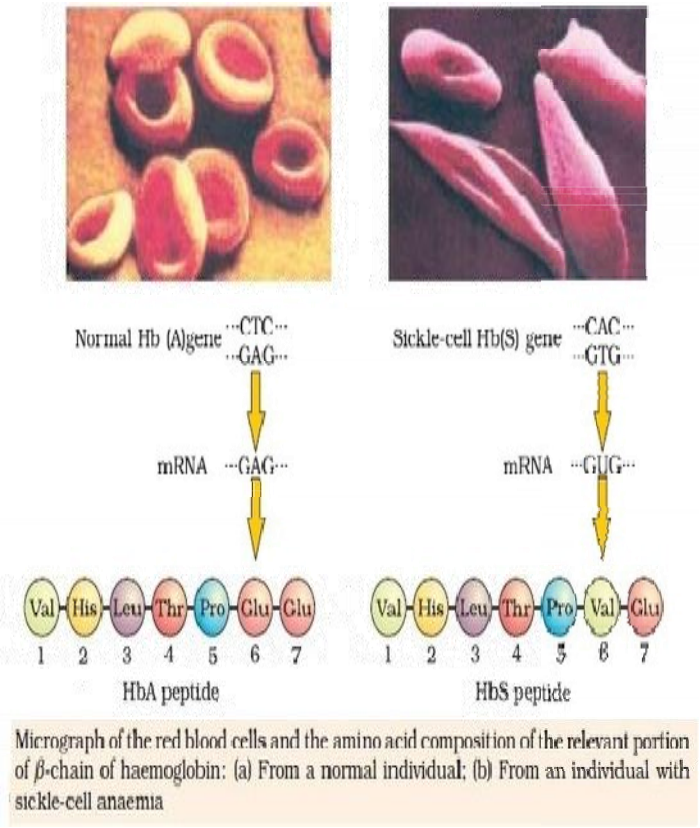
- This is **an autosome linked recessive trait**
- This can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous).
- The disease is controlled by a single pair of allele, Hb^A and Hb^S.

Genotypes are

- Normal $Hb^A Hb^A$
- Un affected Carrier Hb^AHb^S
- Affected Hb^sHb^s

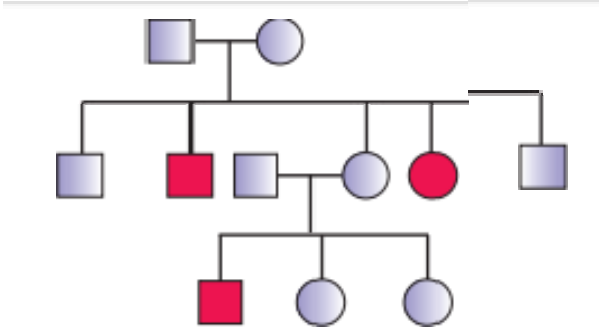
- This disease is transmitted from parents to the offspring when both parents are carrier (Heterozygous)
- **The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule**.

- The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from **GAG to GUG.**
- The mutant haemoglobin molecule undergoes polymerisation **under low oxygen tension** causing the change in the **shape of the RBC from biconcave disc to elongated sickle like structure**



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

Pedigree analysis-Sickle cell anaemia

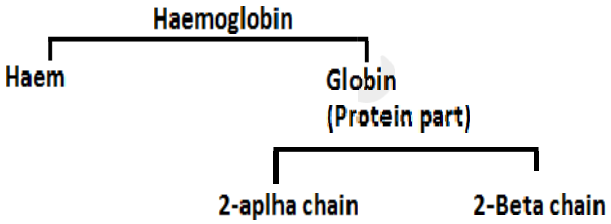
- 

5. THALASSEMIA

- This is the **autosomal linked recessive** trait.:

The genotypes are

- Normal AA
  Carrier Aa
- Affected aa
- This disease is transmitted from parents to the offspring when both parents are carrier (Heterozygous)



- The defect could be due to **either mutation or deletion** which ultimately results in **reduced rate of synthesis of one of the globin chains (α and β chains)** that make up haemoglobin. This causes the formation of **abnormal haemoglobin** molecules resulting into **anaemia** which is characteristic of the disease
- Thalassemia can be classified according to **which chain is affected**
  a) α-thalassemia
  b)β-Thalassemia

a) α-thalassemia

- Here production of **alpha globin** chain is affected
- α-thalassemia is controlled by **2 closely linked gene-HBA-1, and HBA-2**
- These genes are located on the **chromosome number 16 of each parent**
- Mutation or deletion of one or more of the 4 genes results **alpha thalassemia**
- The more gene is affected , less alpha globin molecule is produced

b) β-Thalassemia

- Here production of Beta globin chain is affected

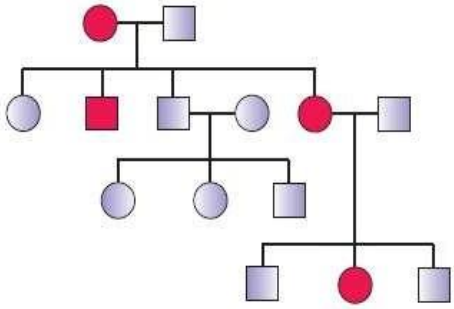
- α-thalassemia is controlled by a single gene **HBB gene.**
- this genes are located on the **chromosome number 11 of each parent**
- Mutation or deletion of one or both genes result Beta thalassemia

Thalassemia differs from sickle-cell anaemia in that the Thalassemia is a **quantitative problem** of synthesizing too few globin molecules while the sickle cell anaemia is a **qualitative problem** of synthesizing an incorrectly functioning globin.

6. MYOTONIC DYSTROPHY

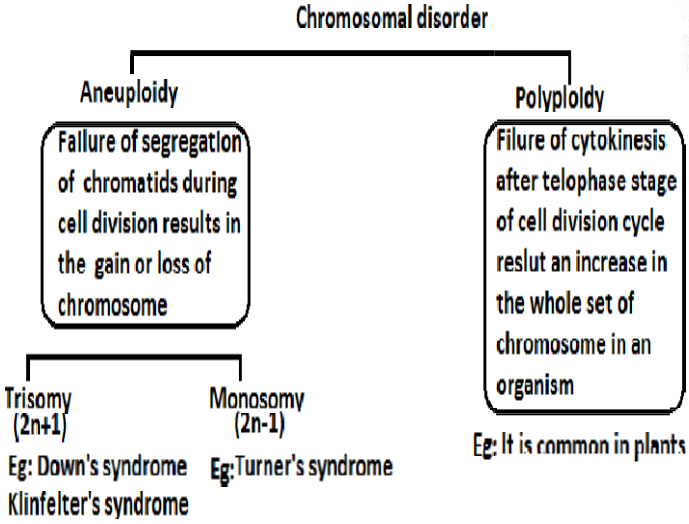
- This is an **autosomal dominant** trait

Pedigree analysis-Myotonic dystrophy



B) chromosomal dissorder

- It is due **to absence or excess or abnormal arrangement of one or more chromosome**



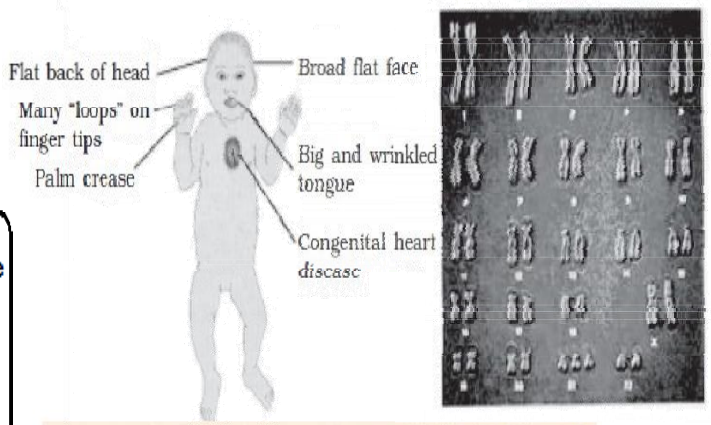
- The total number of chromosomes in a normal human cell is 46 (23 pairs). Out of these 22 pairs are **autosomes** and one pair of chromosomes are **sex chromosome**.
- Sometimes, though rarely, either an **additional copy of a chromosome** may be included in an individual this situation is called **Trisomy** .
- Sometimes though rarely an individual may lack **one of any one pair of chromosomes,** this situation is called **monosomy .**

1. Down's Syndrome(45+XX or 45A+XY)

- This is due to an additional copy of the **chromosome number 21 (trisomy of 21).**
- This disorder was first described by **Langdon Down (1866).**

Symptoms

- The affected individual is
  - ✓ short statured
  - ✓ with small round head,
  - ✓ with furrowed tongue and with partially open mouth
  - ✓ Their Palm is broad with characteristic palm crease.
  - ✓ Physical, psychomotor and mental development is retarded.



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

2. Klinefelter's Syndrome (44A+XXY)

- This genetic disorder is also caused due to **the presence of an additional copy of X-chromosome** resulting into a karyotype of 47, XXY.

Symptom

- Such an individual has overall masculine development, however, the feminine development (development of breast, i.e., **Gynaecomastia**) is also expressed. Such individuals are sterile

Klinfelter's syndrome

3. Turner's Syndrome : (44A+XO)

- It is due **to the absence of one of the X chromosomes,** i.e., 45 with X0,

Symptoms

Such females are

- sterile
- ovaries are rudimentary
- lack of other secondary sexual characters

❖

Turner's syndrome

Concept of Dominance

✓ Every gene contains the information to express a particular trait.

✓ A gene that contains the information for producing an enzyme

✓ In a diploid organism, there are two copies of each gene, i.e., as a pair of alleles.

✓ In heterozygous conditions (Tt), there are dominant and recessive alleles.

Example :

✓ The normal allele (functioning allele or **unmodified allele**)of a gene produce a normal enzyme that is needed for transformation of substrate into product

✓ **The modified allele responsible for production of**

i) Normal or less efficient enzyme

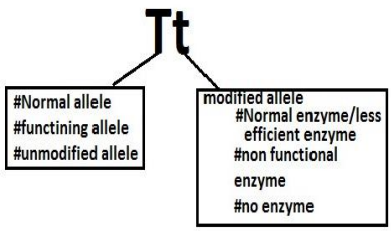
ii) A non functional enzyme

iii) No enzyme at all

❖ **In the first case**, the modified allele produce same phenotype like unmodified allele, so it become dominant

In the 2nd and 3rd case, the phenotype will depend only on the functioning of un modified allele.

Ie:Modified allele become recessive



- Hence, in the heterozygous condition, the recessive trait is seen due to non-functional enzyme or because no enzyme is produced.

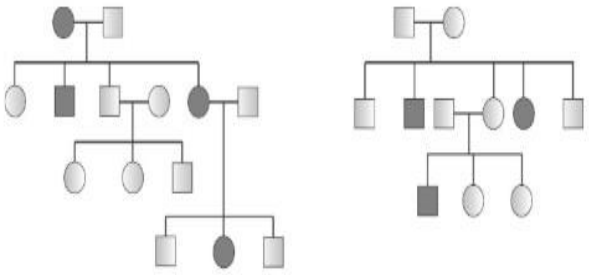
PRINCIPLES OF INHERITANCE AND VARIATION

HSE-March 2024

1. Genes which code for a pair of contrasting traits are known as_____. (1)
2. Complete the following table :

Chromosomal Disorder	Karyotype	Symptoms
A	Trisomy 21	Palm is broad, Short statued
B	C	Gynaecomastia
Turner's Syndrome	44 + Xo	D

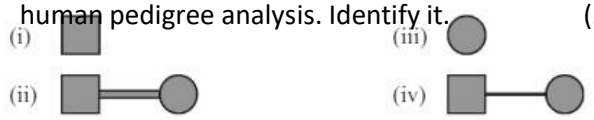
3. In a cross between true breeding red-flowered (RR) and true breeding white-flowered (rr) plants, the F1 (Rr) was pink coloured.
   (a) Name the inheritance pattern mentioned here. (½)
   (b) F1 was self-pollinated and F2 was obtained. What is the genotypic ratio and phenotypic ratio of F2. (1)
   (c) Mention a plant which shows this inheritance. (½)
4. Pedigree analysis of two Mendelian disorders are shown below :



   (a) Identify the trait represented as A and B (1)
   (b) Which is the symbol for consanguineous mating used in pedigree analysis ? (1)
   (c) What do you mean by pedigree analysis ? (1)

HSE-June 2023

5. The tiny insect selected by Thomas Hunt Morgan as his experimental material is _____. (1)
6. "Scientifically it is correct to say that the sex of the baby is determined by the father and not by the mother !" Do you agree with this statement ? Justify your answer (2)
7. Given below are some symbols generally used in human pedigree analysis. Identify it. (2)



8. Match the following : (2)

(a)	Thalassemia	(i)	Gynaecomastia
(b)	Haemophilia	(ii)	Inborn Error of Metabolism
(c)	Down's syndrome	(iii)	Autosome-linked recessive blood disease
(d)	Klinefelter's syndrome	(iv)	Sex linked recessive disease
		(v)	Trisomy 21

HSE-March 2023

9. Match the following (2)

Genetic Disorders	Genetic Reasons
Klinefelter's syndrome	21st Trisomy
Down's syndrome	Lack of one 'X' chromosome in females (XO)
Turner's syndrome	Due to autosomal recessive trait
Phenylketonuria	Presence of an extra X chromosome in males (XXY)

10. Cross between Red flower (RR) and white flower (rr) bearing plants of Snapdragon produced all plants with pink flowers in F1 generation. (3)
    (A) Name the genetic phenomenon of this cross.
    (B) Illustrate F2 generation of this cross using Punnet square

HSE- July 2022 (SAY/IMP.)

11. Various symbols are used in human pedigree analysis. Identify the following symbols (2)

a) 

b) 

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12. TH Morgan carried out several dihybrid crosses in Drosophila. Mention two reasons for selecting Drosophila as an experimental organism? (2)
13. Characters of certain genetic disorders are given below. Identify the given disorders (3)
    a) Sex linked recessive disorder that affect the clotting of blood
    b)The disorder caused by the substitution of Glutamic acid by Valine at the sixth position of beta globin chain of Haemoglobin
    c)The inborn error metabolism and affected individual lacks an enzyme that converts Phenyl alanine to Tyrosine.

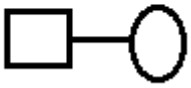
HSE- March 2022

14. (a) Distinguish between Male heterogamety and Female heterogamety. (2)
    (b) Write one example for each.
15. "Sex of a child is determined by father." Substantiate the statement (3)

HSE- August 2021

16. Identify the symbol used in pedigree analysis(1)

a) 

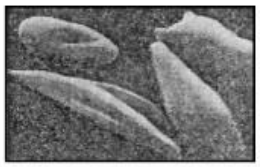
b) 

17. T.H Morgan selected *Drosophila melanogaster* as a suitable experimental organism. Mention any two reason for selecting Drosophila as experimental organism (2)
18. Consider the two statements regarding the haemophilia disorder (2)
    (i) It is an autosome linked dominant disease.
    (ii) The heterozygous female (carrier) for haemophilia may transmit the disease to son. Select the wrong statement and correct it.

HSE-March 2021

19. Name the genetic disorder in which a blood clotting protein is affected leading to non-stop bleeding even through a simple wound. (1)
20. Presence of an additional copy of chromosome 21 was observed in a person during diagnosis. (2)
    (a) Identify the genetic disorder
    (b) Write the characteristic features of this disorder
21. If a father is with 'O' blood group and mother is with 'B' blood group, write the possible blood groups of their children. (2)
22. Micrograph of Red blood cells of two persons (A) and (B) are shown below. Person B is affected with a specific genetic disorder.
    (i) Identify the genetic disorder.
    (ii) Write reason for this disorder.


(A)


(B)

23. 'Incomplete Dominance' is an example for deviation from Mendelian Inheritance. Illustrate with example (3)
24. Consider the two statement regarding the haemophila disorder
    a) It is an autosome linked dominant disease
    b) The heterozygous female (carrier) for haemophilia may transmit the disease to son
    Select the wrong statement and correct it

25. A monohybrid cross is given below :

Parents: Tall X Dwarf

TT tt

Gametes T t

F1 Tt

Find the F2 phenotype and genotype ratio (2)

26. Distinguish male heterogamety and female heterogametywith example (3)

HSE-July-2020

27. Select a female heterogametic animal from the following : (1)
    (a) Human beings (b) Drosophila
    (c) Birds (d) Grasshopper

28. Complete the table using appropriate terms : (2)

Klinefelter's syndrome	(a)	Sterile Male
(b)	44A + XO	(c)
(d)	Trisomy 21	Mental retardation

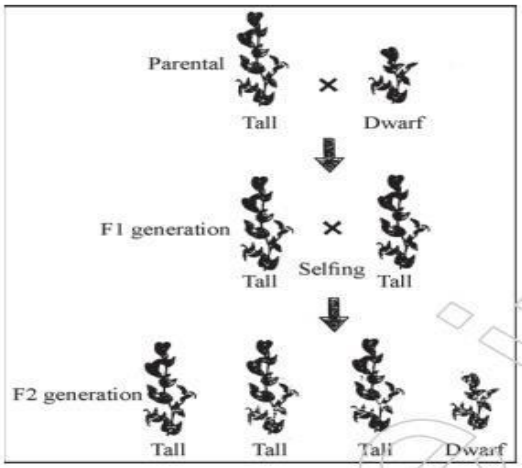
29. In a cross between a true breeding red flowered and a true breeding white floweredplants, the F1 generation was pink coloured flowers. From this cross – (2)
(a) Identify the Inheritance.
(b) Give an example for this type of Inheritance.
(c) Write the F2 phenotypic and genotypic ratio.

HSE-March-2020

30. From the following, find out the symbol used in the human pedigree analysis representing male. (1)

(a) ○ (b) ◇
(c) □ (d) ●

31. Observe the figure given below showing Mendel's experiment using pea plants. (2)
a)

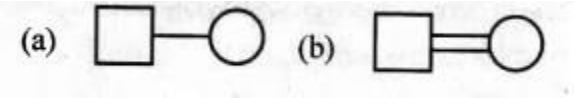


(a) Identify the cross
(b) Which are the laws proposed by Mendel based on this observations ? (2)

32. Correct the following statements, if there is any mistake :
(a) Haemophilia is a autosome linked recessive disease.
(b) Turner's syndrome is due to the presence of an additional copy of X chromosome

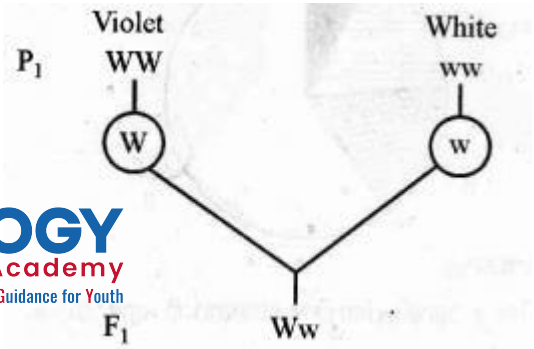
HSE-June-2019

33. Identify the following symbols in pedigree Analysis



(1)

34. Observe the cross of a pure violet and white flower (2)



By using the F, progeny design a test cross.
b) Mention the significance of test cross

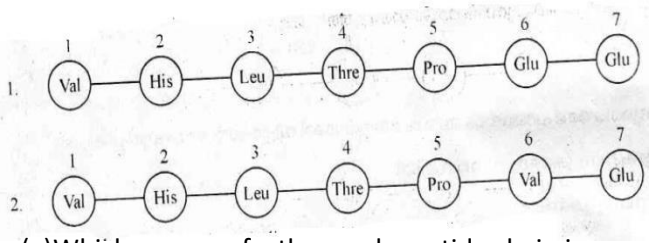
35. Each symptom of two chromosomal disorders are given below : (2)
- Gynaecomastia
- Rudimentary ovary and lack of secondary sexual characters

(a) Identify the disorders.
(b) Give the reason for these disorders

HSE-March-2019

36. Find the odd one out. Justify your answer.
Down's syndrome, Turner's syndrome, phenylketonuria, Klinefelter's syndrome (2)

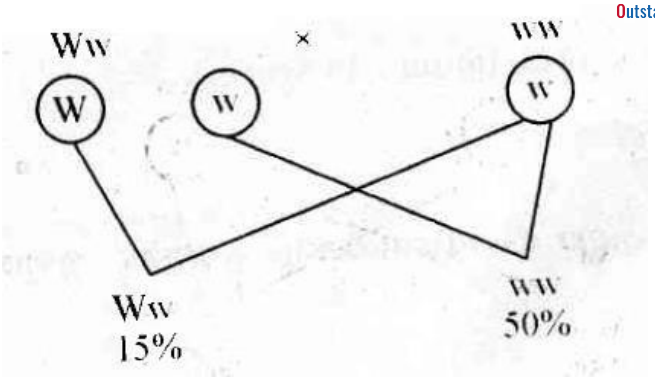
37. The amino acid composition of the relevant portion of β chain of two haemoglobin molecule molecules (A & B) are shown below (3)



(a)Which one of the polypeptide chain is abnormal?
(b) Name the disorder caused by it.
(c) What is the reason for this abnormality?
(d) What is the effect of this abnormality in such individuals?

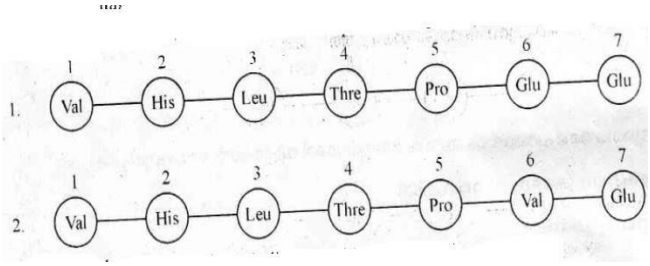
HSE-June-2018

38. Observe the following cross between heterozygous dominant progeny and homozygous recessive parent. Answer the following questions (2)



a) Identify the cross?
b) Mention the significance of this cross?

39. The following diagram shows amino acid sequences of a part of β chain of haemoglobin of 2 individuals. Observe the amino acid sequence and answer the following questions : (2)



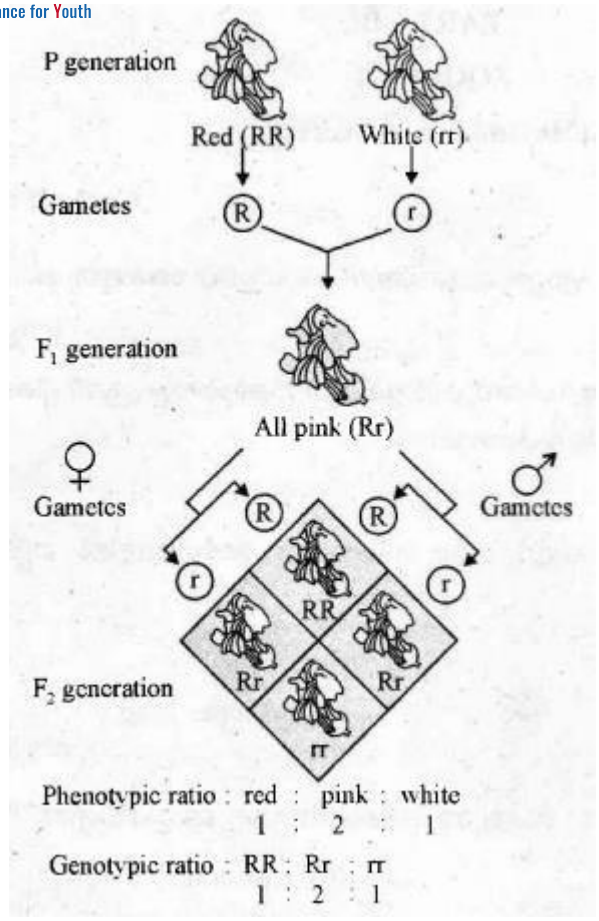
a) Which among the above indicate sickle cell anemic condition?
b) Justify your answer?
c) Describe what is single base substitution?

40. The blood group of a child is 'O'. His father is with 'A' blood group and mother with'B' blood group. Write, down the genotype of the child and genotypes of parents.(2)

HSE-March-2018

41. In a classroom discussion, a student said that the sex of the baby is determined by father. Analyze the statement and give reason for it ? (2)

42.



a) Observe the above cross and name this phenomenon?
b) Write down the theoretically given explanation of the phenomenon (2)

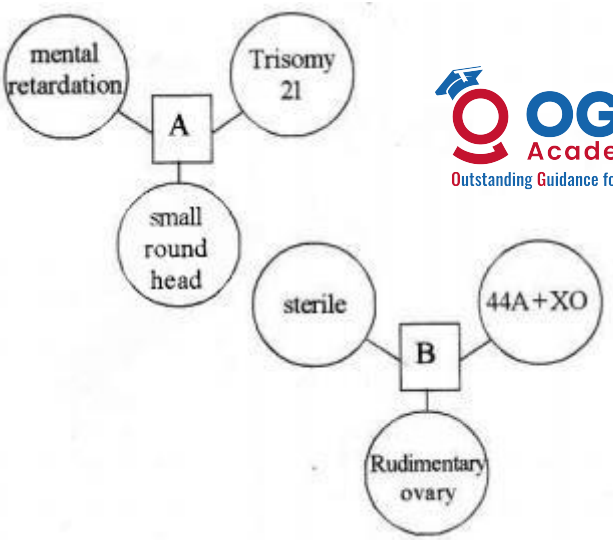
43. Haemophilia, Sickle cell anaemia and Phenyl Ketonurea are Mendelian disorders
(a) What do you mean by mendelian disorder
(b) which one of the above is an example of in born error of metabolism? Mention the cause of disorder? (2)

HSE-Model Exam -2018

44. Construct a monohybrid cross between homozygous violet and white coloured flowers of a pea plant How can one determine whether the F1 Progenies are homozygous or heterozygous? (2)

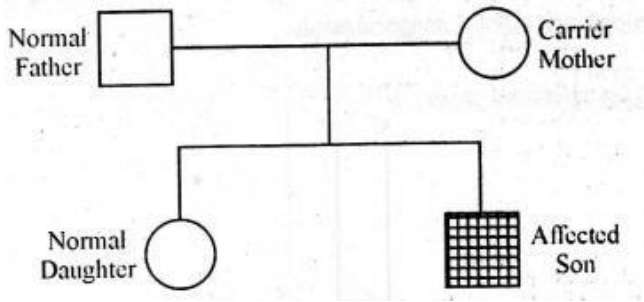
45. From a clinical laboratory, Ramu's blood group was identified as 'AB' goup. But his father has 'A' blood group and mother has is 'B' blood group.
a) Is Ramu's blood group identification correct?
b) Substantiate your answer using co dominance principle. (2)

46. Identify the syndromes 'A' and 'B' (2)



HSE-JUNE-2017

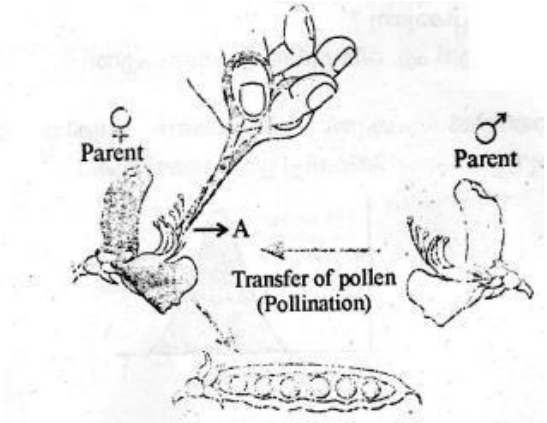
47. Observe the diagrammatic representation of following pedigree analysis and answer the question. (3)



a) Describe the type of inheritance shown in the diagram

b) Distinguish between Mendelian disorder and chromosomal disorder with example?

48. Observe the following diagram and answer the question
(Hint: step in making a cross in pea plant) (2)



a) Name the process marked as A and writes its significance?

b) Diagrammatically represent a monohybrid cross between Tall and dwarf pea plant

HSE-MARCH-2017

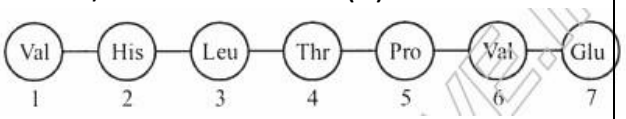
49. The following table shows the F2 generation of a Dihybrid cross. Identify the phenotype with homozygous recessive genotype. Find out A:B:C:D (2)

No.	Phenotype	No. of offspring (F_2 gen.)
1	A	21
2	B	7
3	C	63
4	D	21

50. Which of the following do not have similar sex chromosome? (homogametic) (1)

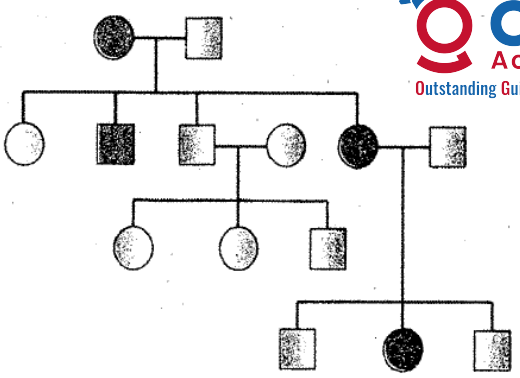
(1) Human female
(2) Drosophila female
(3) Bird female
(4) Bird male

51. Examine the following fragment of beta globin chain in human haemoglobin and identify the hereditary disease with reason(2)



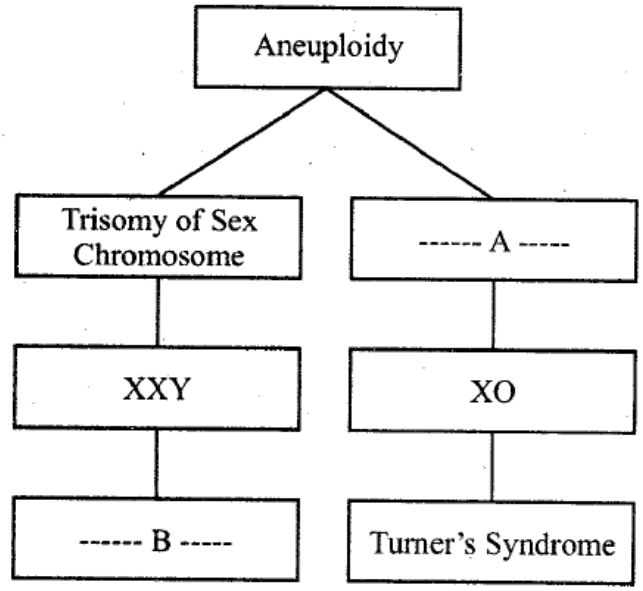
HSE-June-2016

52. Observe the figure below and answer the question following : (2)



a) Identify the figure?
b) what show the shaded symbols used?

53. a)Complete the flow chart of chromosomal disorder by filling the blank boxes (A and B) (3)



b)What is aneuploidy ?

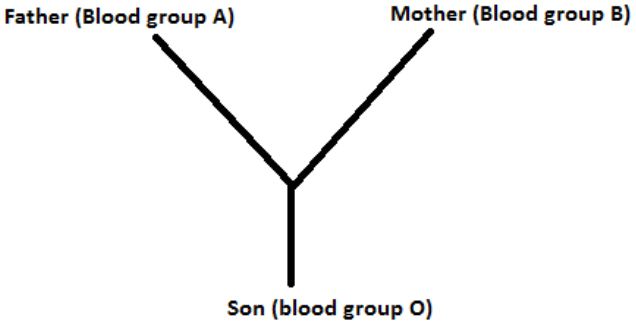
HSE-March-2016

54. Which of the following is not a Mendeliandisorder (1)

Colourblindness, Down's syndrome, Haemophilia, Thalassemia

55. Study the following cross and answer the questions.

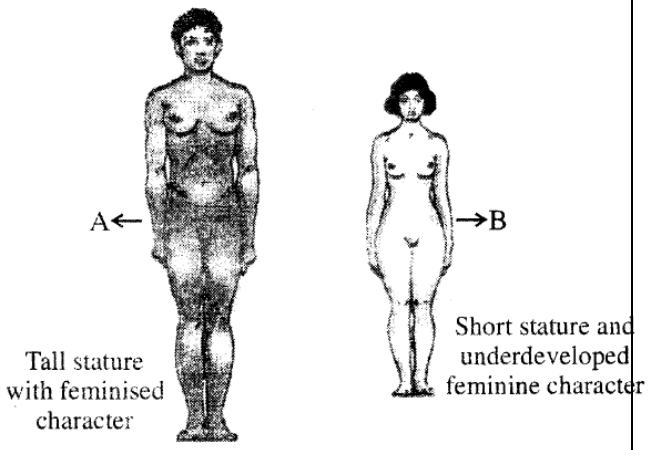
[Hint: ABO blood group in man is controlled by three alleles I^A, I^B and i.]



a) Write the genotypes of Father, Mother and Son.

b) The type of dominance of human blood group inheritance is..... (2)

56. Observe the figure and answer the question (2)

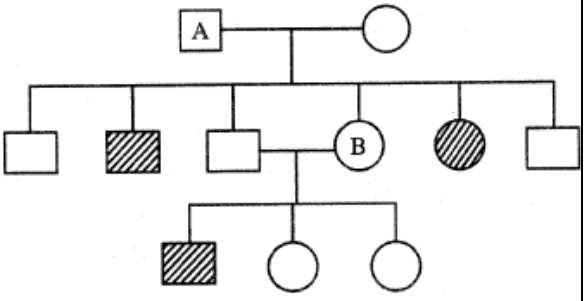


a) Identify the syndromes A and B.?

b) What is the chromosome numbers in A and B?

HSE-SAY-2015

57. Diagrammatic representation of the pedigree analysis of the inheritance of sickle cell anaemia is shown below. (3)



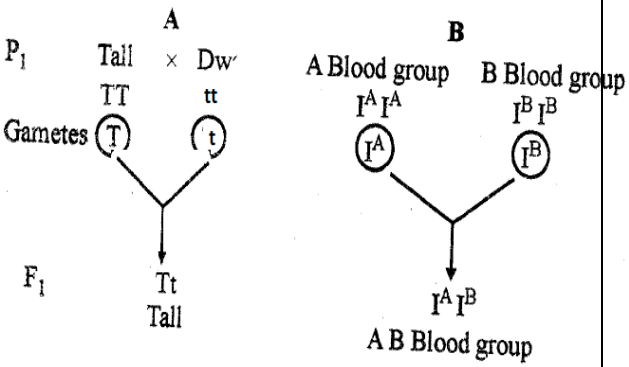
a) Name the type of inheritance shown in the figure ?

b) Write the genotype of A and B?

(Hint : Disease is controlled by a pair of allele $Hb^A$and Hb^s)

c) Represent pedigree analysis of an X linked Recessive Inheritance diagrammatically

58. Observe the inheritance shown in A and B

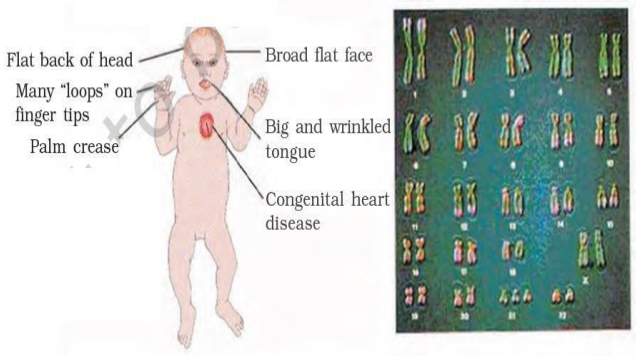


a) Name the type of inheritance shown in A and B?

b) What is the difference between the two types of inheritance? (2)

HSE-March-2015

59.



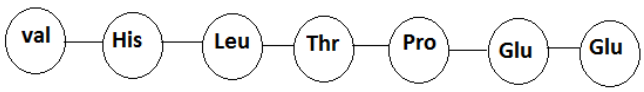
a) Identify the syndrome from the diagram, and write the genotype?
b) It occurs in both sexes (Male and female)? Write the reason (2)

60. Fill in the blanks: (1)
a). is a metabolic disorder that occurs due to the lack of an enzyme that converts phenyl alanine to tyrosine.
b). is a disease caused by the substitution of glutamic acid by valine at the 6th position

61. It is evident that, it is the genetic make of a sperm that determine the sex of the child in human beings. Substantiate (2)

HSE-SAY-2014

62. Correct the amino acid sequence of sickle cell hamemoglobin (1)



63. Identify they syndrome from the genotype given below: (1)
a) 44 Autosome + XXY
b) 44 Autosome +XO

64. Sex of the Baby is determined by the father, not by the mother. Substantiate (2)

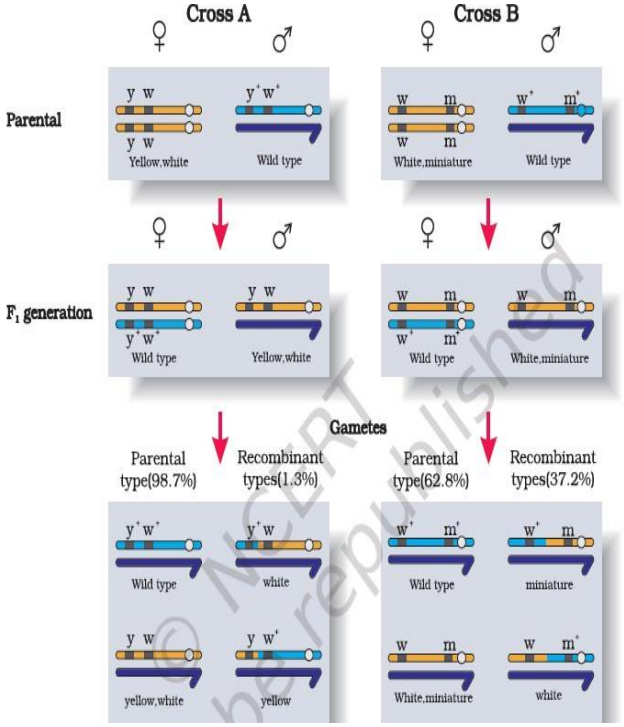
65. a) Define mutation (1)
b) What are the different types of mutation? (1)

66. The family of Queen Victoria shows a number of Haemophilic descendants as she was the carrier of the disease. Name the pattern of inheritance of this Royal disease. (1)

67. a) Paternity or maternity can be determined by certain scientific methods. What is it? Define
b) Briefly write the methodology involved in the technique ?
c) comment on its other application (3)

HSE-March-2014

68. Explain the phenomenon shown in the following figure and the reason for difference in the production of recombinant in Cross A and cross B as explained by Morgan. (3)

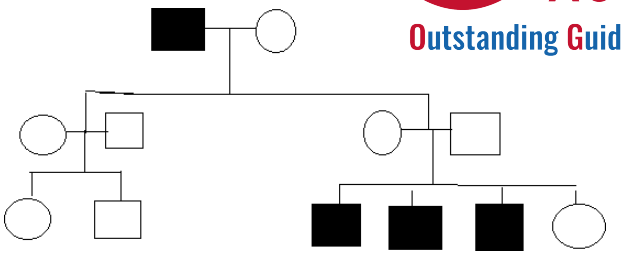


69. Difference in chromosome number of some human being A,B,C, and D is given below:
A) 22 pairs of Autosome
B) 22 pairs of Autosome +XO
C) 22 pairs of Autosome+ 1 autosome
D) 22 pairs of Autosome+ XXY
a) Identify the person with who suffers from Klinfelter's syndrome. Write its symptoms
b) Differentiate between aneuploidy and polyploidy ? (3)

70. Gopalan argues that if the father is of 'A' blood group, Mother is of 'B' blood group. Their children can be only be 'A' group, 'B' group or 'AB' group.
a) Do you agree with Gopalan'sarguement?
b) Give reason for your argument? (2)

HSE-SAY-2013

71. In the given pedigree the shaded figure denotes individuals expressing a specific trait (2)



Which of the following is the most probable mode of inheritance of this trait

A-Simple mendalian recessive inheritance

B-Co dominant Relationship of a single pair of allele

C-X linked recessive transmission

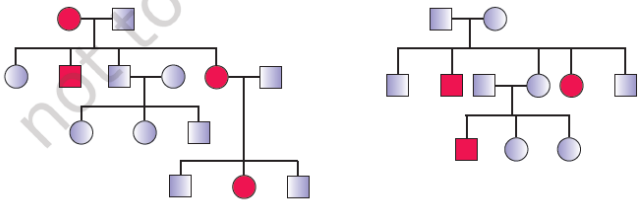
D-X linked dominant transmission

E-Polygenic inheritance

HSE-March-2013

72. Identify the trait from pedigree chart. Give one example each. (2)

a)

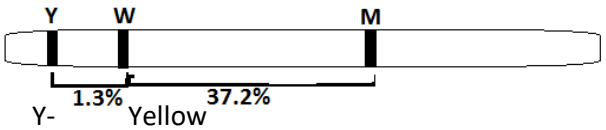


(a) (b)

73. A poultry farm manager was cursing his hens for producing lion share of cocks in its progeny. Hearing this, Kumar-farm manager starts to lame his wife for delivering consecutive girl children. Analyse the situation scientifically and state whether you agree with kumar? (3)

HSE-SAY-2012

74. Diagrammatic representation of chromosome map of Drosophila is given below (2)



Y- Yellow

W- White

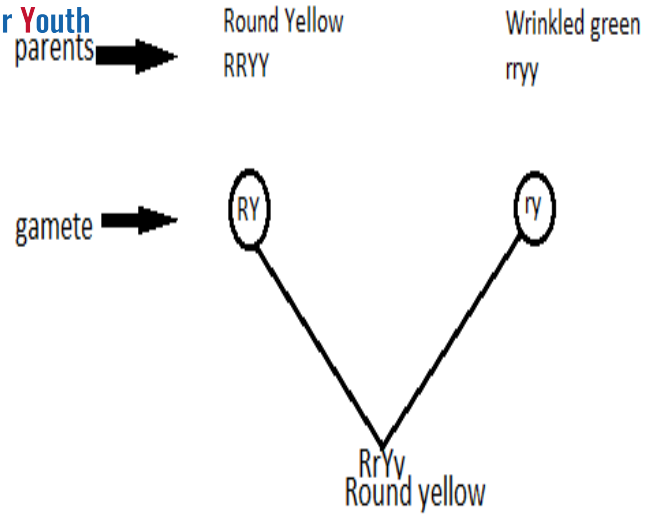
M- Miniature

a) Which genes are more linked?

b) Who mapped chromosome firstly?

c) Tightly linked genes show low recombination. Why?

75. Work of a student is given below: (3)



From the above give an example for genotype and phenotype?

b) Complete the work using the punnet square and find out the phenotypic ratio in the F2 generation?

HSE-March-2012

76. Complete the tale using suitable term (2)

Turner's syndrome	.....a.....	Sterile female
---------b--------	44A+XXY	.....c.....
--------d---------	Trisomy-21	Mental retardation

77. In Pea plant the gene for yellow seed colour is dominant over green and round seed shape is dominant over wrinkled. Write the four types of gametes formed in heterozygous pea plant with Yellow and round seeds (YyRr) (1)

78. The first child of a couple is affected with Phenyketonuria. During the second pregnancy they visited a genetic counsellor and Prepared a pedigree chart of their family. (2)

a) What is pedigree analysis?

b) Draw the symbols for

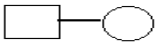
i) Affected female

ii) Sex unspecified

iii) Consanguineous mating

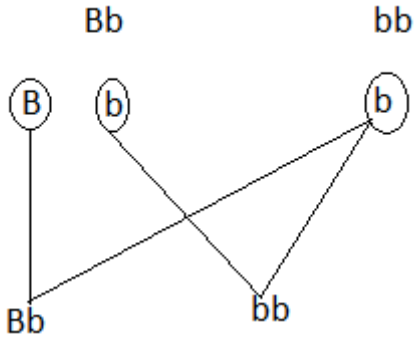
HSE-say-2011

79. Symbols used in human pedigree analysis and their meanings are provided in the table. Fill in the blanks with suitable meaning or symbols (1)

symbols	Meaning
	...a.....
.....b.....	female
	mating
	...c.....
....d.....	affected male

80. Certain facts related to human disorder are given:
1)It is inborn error in metabolism
2)It is inherited as an autosomal recessive trait
3)The affected person is mentally retarded
a)name the disorder
b)What are the physiological processes behind this mental retardation (2)

81. A genetic cross is represented below (2)



a) Identify the given cross?
b) Elaborate upon the significance of such cross?

HSE-March-2011

82. The frequency of occurring Royal disease or Haemophilia is high in the pedigree of Royal families of Queen Victoria. Which of the following cannot be generally inferred from this? (1)
a)Queen Victoria was not homozygous for the disease
b)Many heterozygous families were there in the Royal family
c)Non-Royal families were not affected with haemophilia
d)There is less possibility to become a female diseased
e)Generally a diseased female cannot survive after the first menstruation
f)Pedigree analysis is the study of inheritance patterns of traits in human female.

83. After analyzing the karyotype of a short statured Round headed person with mental retardation, a general physician noticed an addition of autosomal chromosome .
Answer the following question (2)
a)Addition or deletion of chromosome generally result in.....
b)What may be the possible syndrome or disorder of the above person should suspected to be?
c)Suggest two or more morphological peculiarity to confirm the chromosome disorder in that person?

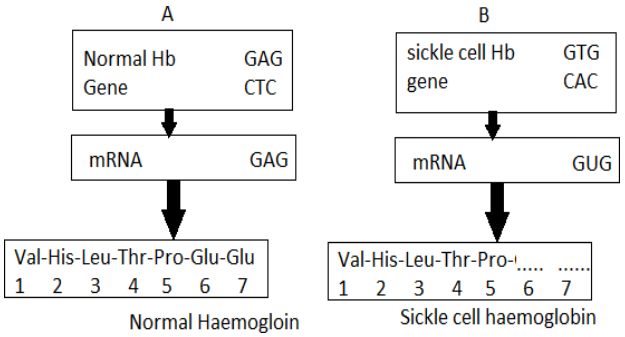
84. A couple has 2 daughters. The blood group of husband and wife is O (2)
a) What is possible blood groups of the children should have?
b) Whether any change in blood group will occur if they have two sons instead of daughters?

HSE-SAY-2010

85. Some genetic abnormalities, their genotype and features are distributed in Column A,B and C respectively . Match them correctly (1.5 mark)

Column A	Column B	Column B
Down's syndrome	44A+XO	Rudimentary ovary and sterility
Turner's syndrome	44A+XXY	Furrowed tongue and partially opened mouth
Klinfelter's syndrome	45A+XX/XY	Gynaecomastia and sterility

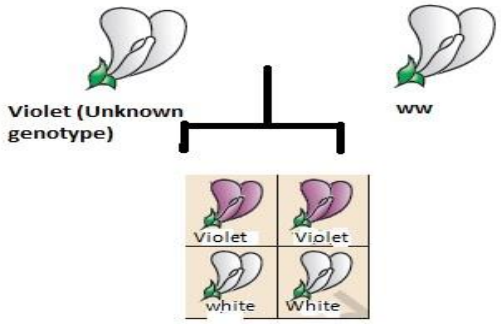
86. The flow chart A and B given below represents the inheritance of normal haemoglobin and sickle cell haemoglobin (3.5)



a) Observe the Flow chart A and complete the flow chart B
b) Note down the genotype of a sickle cell anaemia patient and mention the symptom of the disease
c) Mention the peculiarity of Hb^AHB^s phenotype

HSE-March-2010

87. To findout the unknown genotype of a violet flowered pea plant a researcher done the flowering cross. Observe the diagram and answer the following question:
(Hint :Violet flower colour in pea plant is dominant over white)



a) What would be the above cross called?
b) can you determine the unknown genotype of violet flowered parent by drawing Punnet square?

88. Polypeptide chains of two haemoglobin molecules are shown below. One of the chains shows an abnormality. Observe the diagram and answer the following questions

a) Which of the polypeptide chain in the haemoglobin is abnormal leading to a disease?
b)What is the reason for this abnormality ?
c)What will be the effect of this change in polypeptide chain ?