PRINCIPLES OF INHERITENCE AND VARIATION

Genetics

- It is the study of **inheritance and variation**
- It is the study of **genes and chromosomes**

Inheritance

- It is the transmission of characters from **parents to offspring**
- It 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.
  Eg: Sahiwal cow (Punjab)

GENETIC TERMS

- **Allele :**
  They are alternative form of a gene
  Eg: T,t,R,r,Y,y
- **Phenotype**
  The physical appearance of an organism is called Phenotype
  The visible character of an organism is called phenotype
  Eg: Tall plant, blue eye, round seed
- **Genotype**
  The complete genetic constitution of an organism 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 gene
  Eg:Tt,Rr,Yy

GREGOR JOHANN MENDEL

- He was an Austrian monk
- He is known as father of genetics
- He conducted **hybridization experiment** on **garden pea plant (*Pisum sativum*)** for 7 years (**1856-1863**)
- Based on his experiment he proposed **'laws of inheritance'** in living organisms .
- Mendel selected 7 pairs of true breeding plants varieties, which are given below

Sl No.	Contrasting character	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

Reason for selecting garden pea plant

1. Plants shows clear contrasting character
2. Easy to cultivate
3. It has bisexual flower
4. Cross pollination is easy if self pollination is prevented
5. Floral structures are suitable for cross pollination if self pollination is prevented.

Reason for mendel's success

1. Gradual planning
2. Attenetion was focused only on one character at a time
3. Maintenance of accurate record of result obtained
4. Careful experimentation and observation
5. He was a lucky person (didn't find linkage phenomenon)

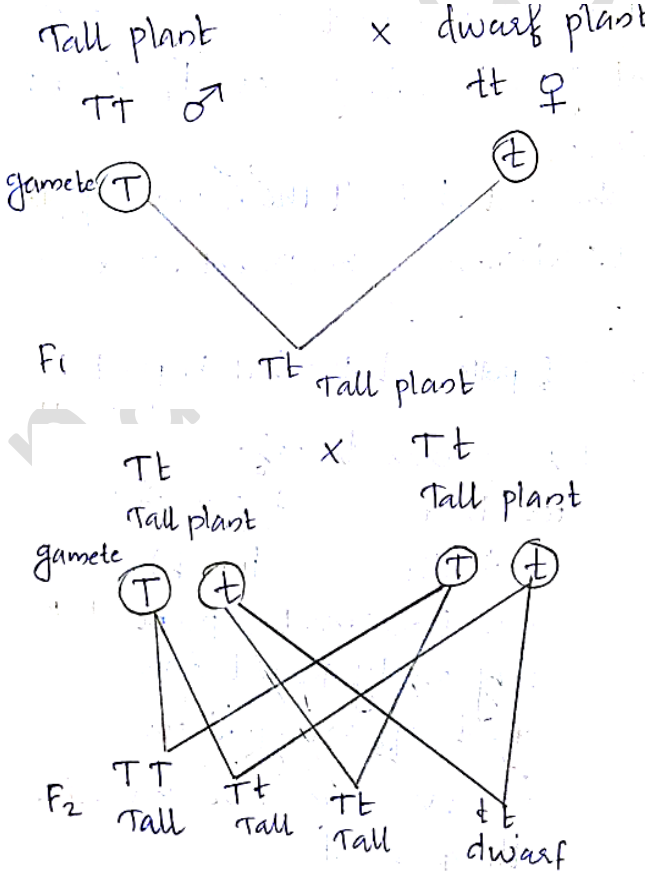
Monohybrid cross-Inheritance of one gene

- It is the cross between 2 parents differing in one character pair.
- 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 (1st filial generation).
- Mendel observed that all the F1 progenies were tall (Like one of its parent and none were dwarf) .

  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)**

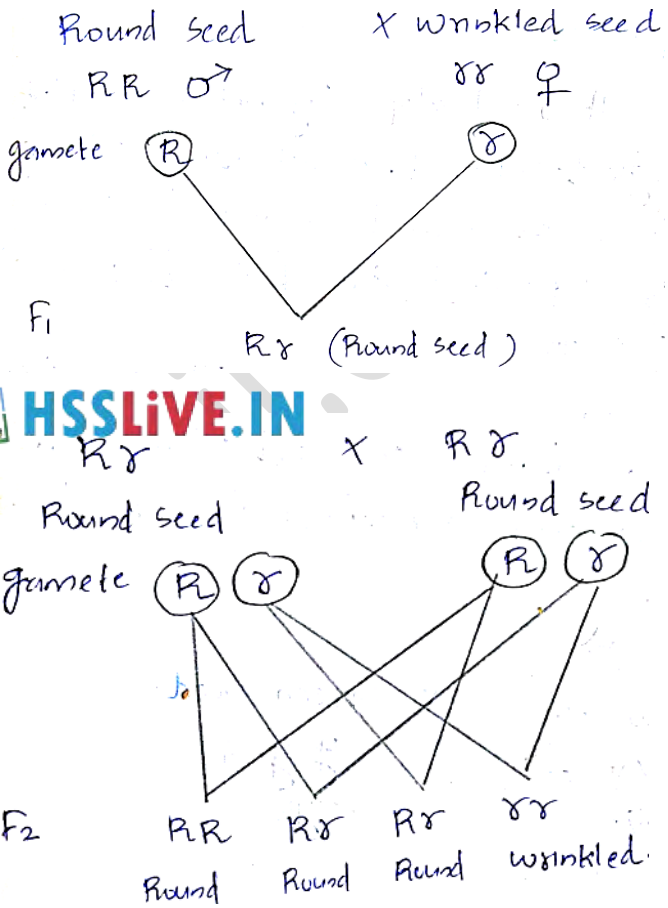
  Ie: characters that was not seen in the F1 generation expressed in the F2 (dwarf)
- Based on this observation mendel proposed that something being was being stably passed down unchanged from parents to offspring through gametes over successive generation. Mendel called it as **factors.** Now we called them as genes.

Example-1



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

Example-2

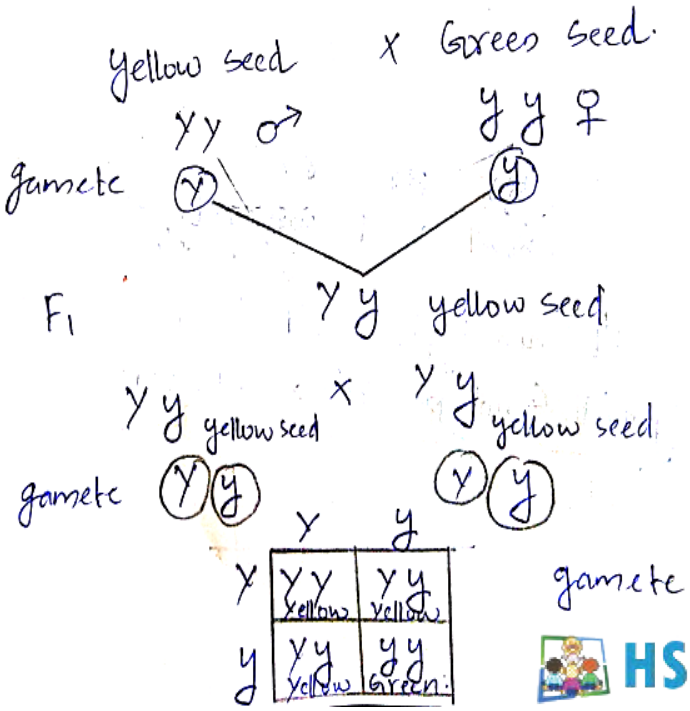


Monohybrid genotypic ratio= 1:2:1
Monohybrid phenotypic ratio =3:1

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.

Example

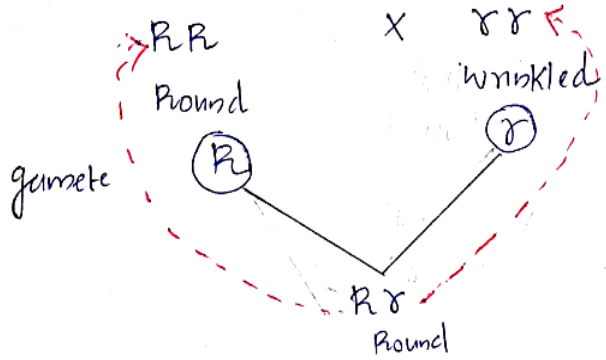


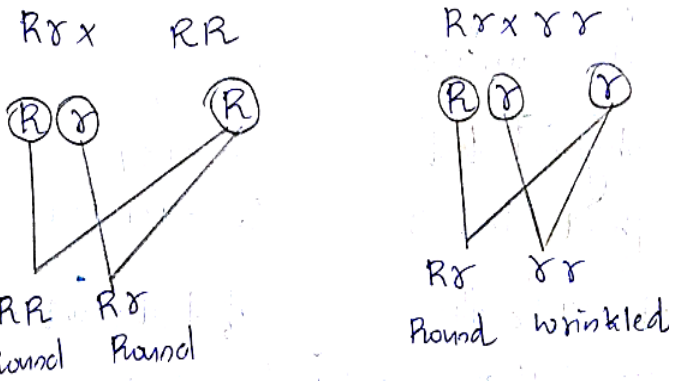
Monohybrid genotypic ratio= 1:2:1
Monohybrid phenotypic ratio =3:1

BACK CROSS & TEST CROSS

Back cross

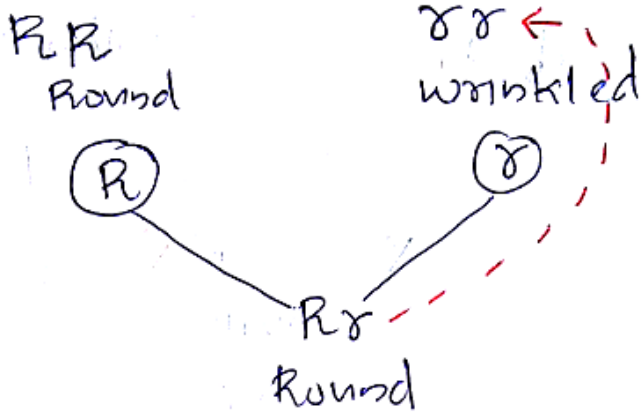
- It is the cross of F1 progeny with **one of its** parent

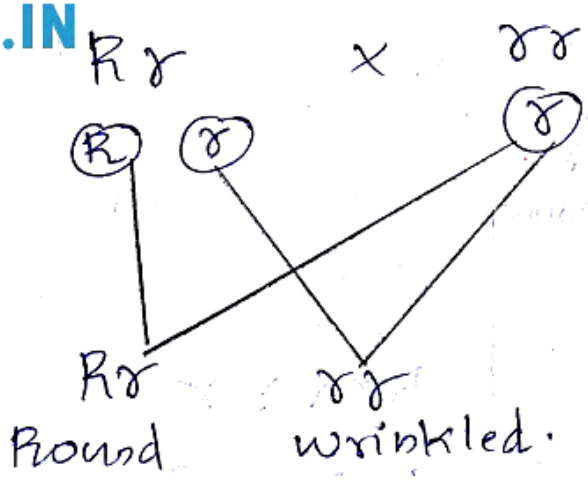




Test Cross

- 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 which produce tall plant and dwarf plant in the ratio 1:1 during test cross.?

Ans:

Mendel's Laws on Inheritance (Principles of inheritance)

1. Law Of Dominance (1st law)

The main points are ...

i) The characters are controlled by discrete units called factors

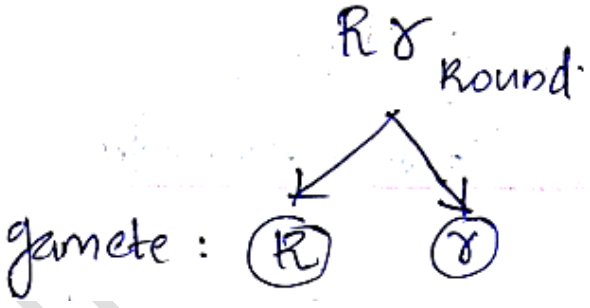
ii) Factors occur in pair

iii)in a dissimilar pairs of factors (Heterozygous) one member of pairs dominates over the other. (The dominated one is called Dominant , and suppressed 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.
- This law explains the proportion of 3:1 obtained at the F2

2. Law of segregation (2nd law/law of purity of gamete)

"During gamete formation 2 factors for a character present in an individual will separate from each other and enter into each gamete"

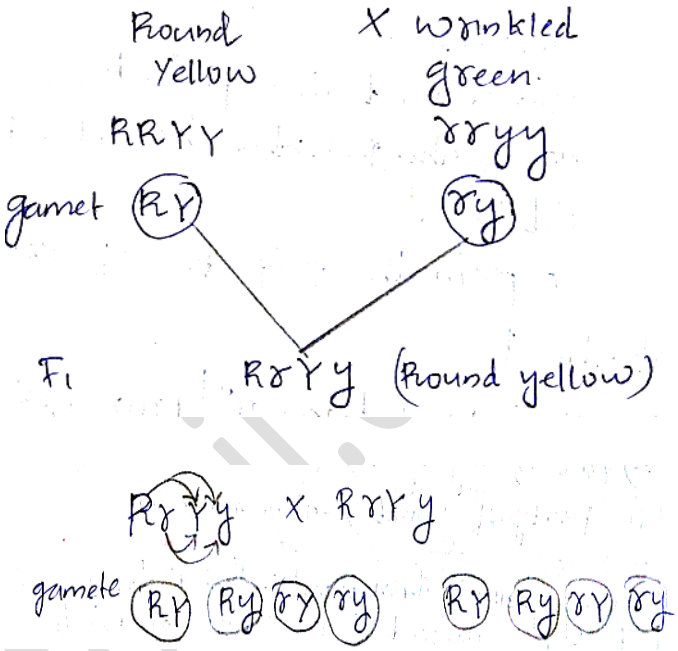


- Both 1st and 2nd law of Mendel obtained from monohybrid cross .

Dihybrid cross-Inheritance of 2 genes

"It is a cross between 2 parents differing in two character pair"

Example-1



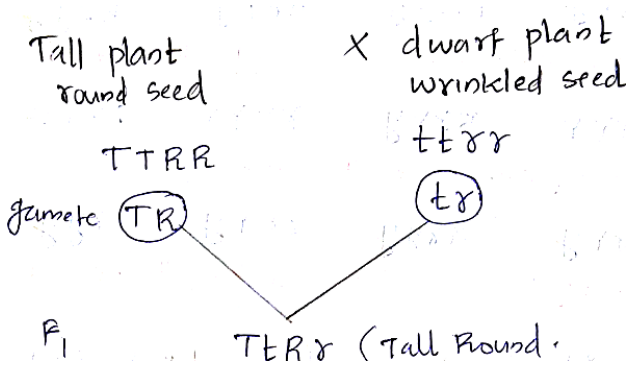
F2.

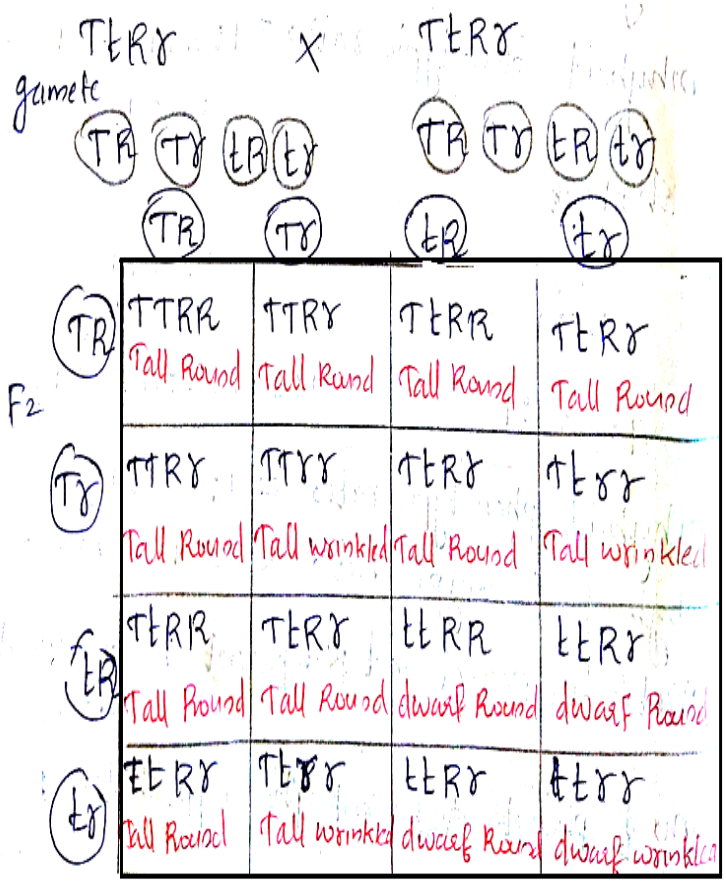
	RY	Ry	rY	ry
RY	RRYY Round yellow	RRYy Round, yellow	RrYY Round yellow	RrYy Round yellow
Ry	RRYy Round yellow	RRyy Round Green	RrYy Round yellow	Rryy Round Green
rY	RrYY Round yellow	RrYy Round yellow	rrYY wrinkled yellow	rrYy wrinkled yellow
ry	RrYy Round yellow	Rryy Round Green	rrYy wrinkled yellow	rryy wrinkled Green.

Dihybrid phenotypic ratio=9:3:3:1

Dihybrid genotypic ratio :1:2:1:2:4:2:1:2:1

Example-2





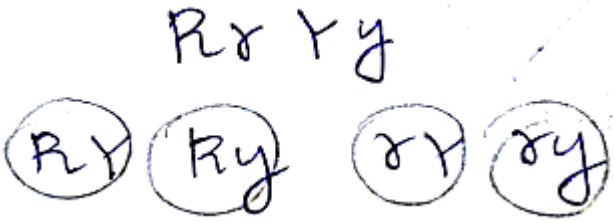
- **Dihybrid phenotypic ratio=9:3:3:1**
- **Dihybrid genotypic ratio =1:2:1:2:4:2:1:2:1**
- **Ratio b/w parent and none parent (recombinant) =10:6**
- **Number of different phenotype in the F2 dihybrid cross = 4**
- **Number of different Genotype in the F2 dihybrid cross = 9**

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

Ans:

3. Law of Independent assortment (3rd law)

- This law is formulated from Dihybrid cross
- This law states that
  "when more than one pair of characters are involved in a cross, factor pairs independently segregate from other pair of characters "
  Ie:



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

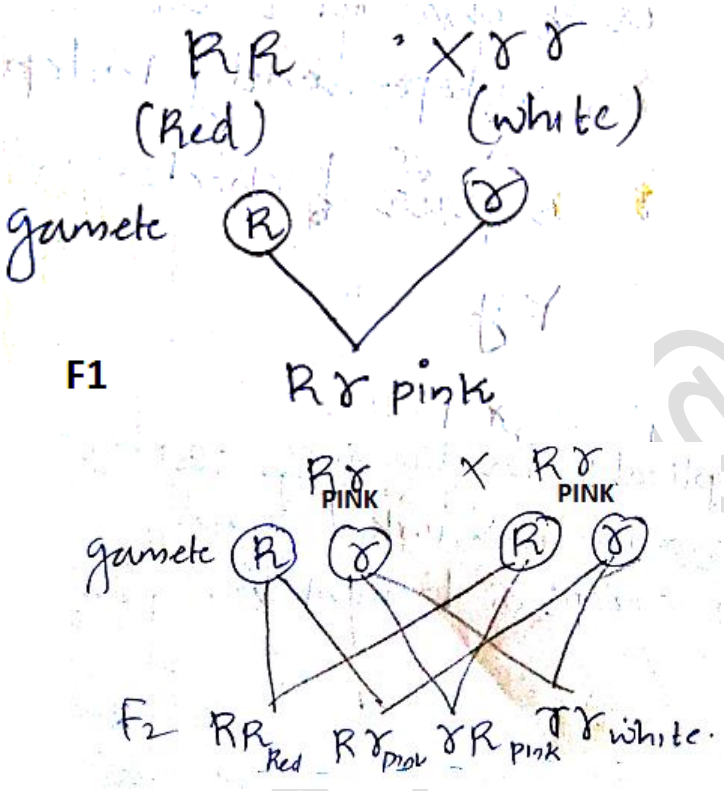
DEVIATION FROM MENDELISM /MENDLIAN PRINCIPLE

1. Incomplete dominance

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

Example-1

- Carl Correns of Germany conducted hybridization experiment in ***Mirabilis jalapa*** **(4 o' clock plant/ snap dragon/Dog flower)** is given below



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

- **In the above example 'R' is not completely dominate over 'r'. This is due to incomplete dominance. So the heterozygoys off springs** show sintermediate character between 2 parent s.

Example-2

Starch grain size in Pea seed

- Starch grain size in pea seed is controlled by one gene. It has 2 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.

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 phenotypes present in ABO blood group

3. Multiple alleles

- Some genes have **more than 2 alleles**. This phenomenon is called multiple allelism .
- **Example-1**

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. Pleotropy

- Multiple effect of a gene is called **pleotropy**
- **Here single gene may produce more than one effect.**
- The underlying mechanism of Pleotropy in most case is the effect of a gene on metabolic pathway-which produce different phenotype.

Example-1

- White eye mutation in Drosophila result changes in body color (depigmentation in various body parts)

Example-2

- Starch grain size in pea seed and seed size

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. it result mental retardation, reduction in hair, pigmentation in skin.

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 .**

Example-1

Human Height

Example-2

Human Skin Colour

- Skin color is controlled by 3 pairs of gene (A,B,C)
- AABBCC--→Dark Skin
- AaBbCc--→Intermediate Color skin
- Aabbcc--→Light color skin

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

i. Communication was not easy
ii. He used maths to explain biological phenomenon was totally new
iii. He could not provide any physical proof for the existence of factors
iv. His concept of factors (Genes) are stable and discrete units was not accepted at that times, because variations are seen in nature .

Rediscovery of Mendelian Principle

- In 1900, three scientist namely **Carl correns, Hugo De Vries, Von Tschermrk independently rediscovered mendel's law in inheritance**
- 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 see 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
- "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
  - Chromosomes are vehicles of heredity
  - 2 identical chromosome forms a homologous pair
  - Homologous pair segregate during gamete formation
  - Independent pair segregate independent each other
  - Behavior of chromosome is parallel to behavior of genes, because genes are located on chromosomes.

A Comparison between the Behaviour of Chromosomes and Genes

Chromosome	Genes
Occur in pairs	*Occur in pairs*
Segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete	*Segregate at gamete formation and only one of each pair is transmitted to a gamete*
Independent pairs segregate independently of each other	*One pair segregates independently of another pair*

Experimental verification of chromosomal theory of inheritance

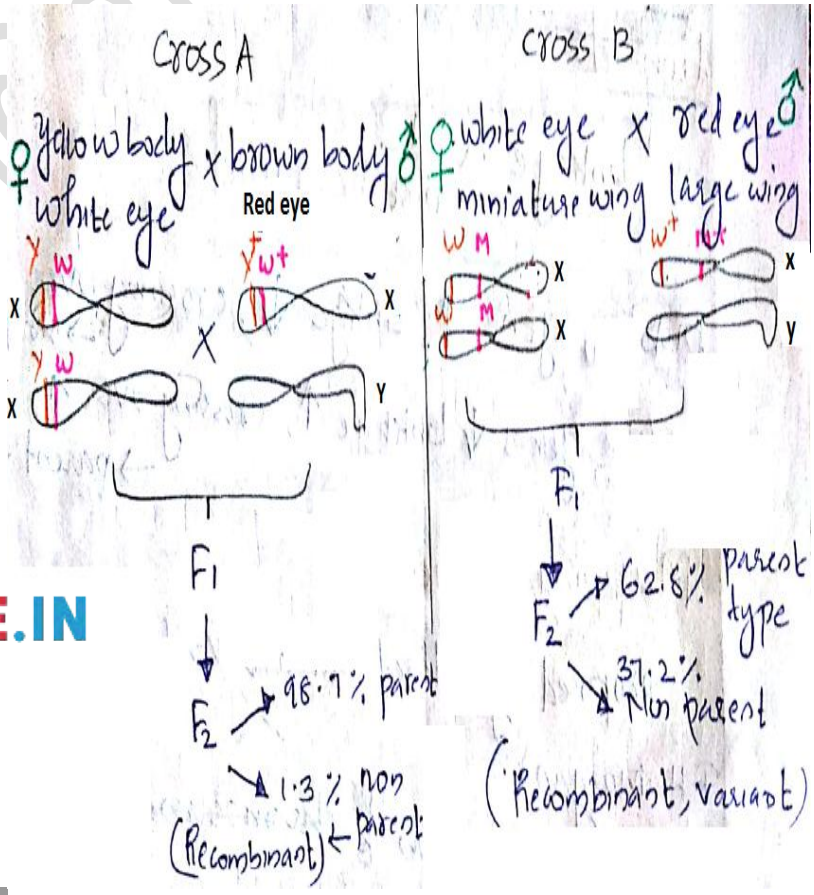
- It is done by **Thomas Hunt Morgan**
- He conducted his experiment of **Fruit fly (*Drossophila melanogaster*)**

Reason for selecting fruit fly

i. It can grown on simple synthetic medium.
ii. It has short life span (2 weeks)
iii. A single mating produce large number of progenies.
iv. There is clear difference between male and female. (females are larger than male).
v. Hereditary variation can seen with low power microscope .

Experiment

T.H. Morgan conducted dihybrid cross to study the genes that were sex linked.



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

- **<u>The physical association of genes in a chromosome is called linkage, such genes are called Linked genes .</u>**
- 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**

<u>Mapping of genes</u>

- Proposed **by Alfred Sturtevant (Student of T.H Morgan)**
- He mapped position of genes in a chromosome
- He used recombination frequency between gen pairs for measuring the distance between genes
- Genetic maps are used in human genome project (HGP)

6. Linkage

- The physical association of genes in a chromosome is called linkage, such genes are called Linked genes .
- Linked genes are **exception to law of independent assortment (3Rd law)**
- <u>The term linkage and crossing over is introduced by TH Morgan</u>.

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

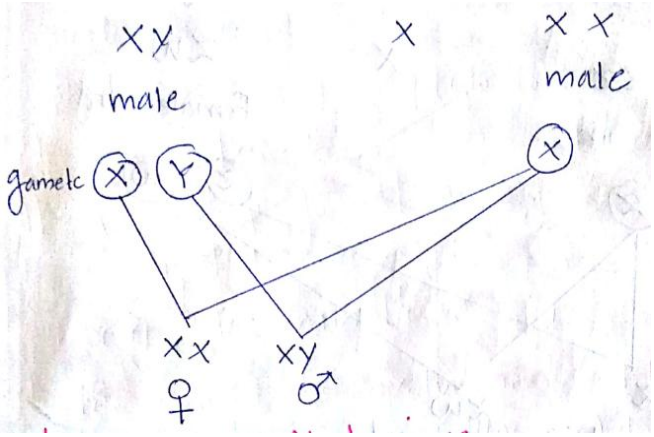
- **Henking (1891)** studied spermatogenesis **in some insects**.
- He observed that 50% of sperm received a nuclear structure after spermatogenesis, other 50% of sperm did not received it .
- Henking called these nuclear structure **as 'X body'**, now it is called as X-chromosome

<u>Mechanism of sex determination</u>

- 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

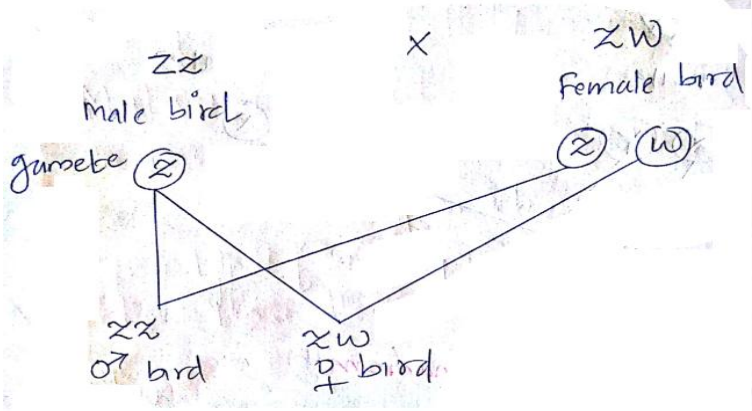
<u>a)XX-XY mechanism –Human being, drosophila</u>

- Here both male and female have same number of chromosomes.
- Males are heterozygous (XY) and produce 2 types of gametes (Sperms) containing only 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

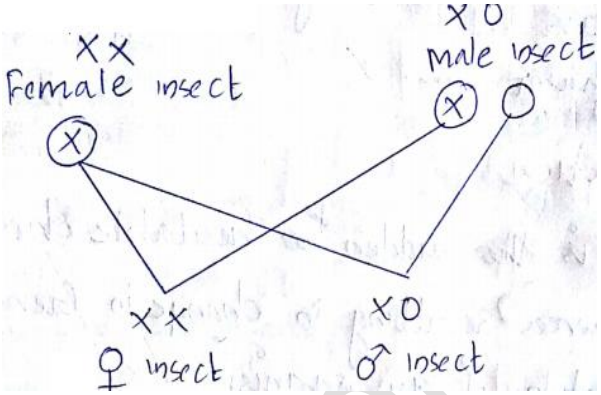
- Here both male and female have same number of chromosomes.
- Females are heterozygous (ZW) and produce 2 types of gametes (Eggs) containing only Z or W chromosomes besides Autosomes.
- Males are homozygoys (ZZ) and produce only one type of gamete (Sperms) containing only Z chromosomes besides Autosomes.
- 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.



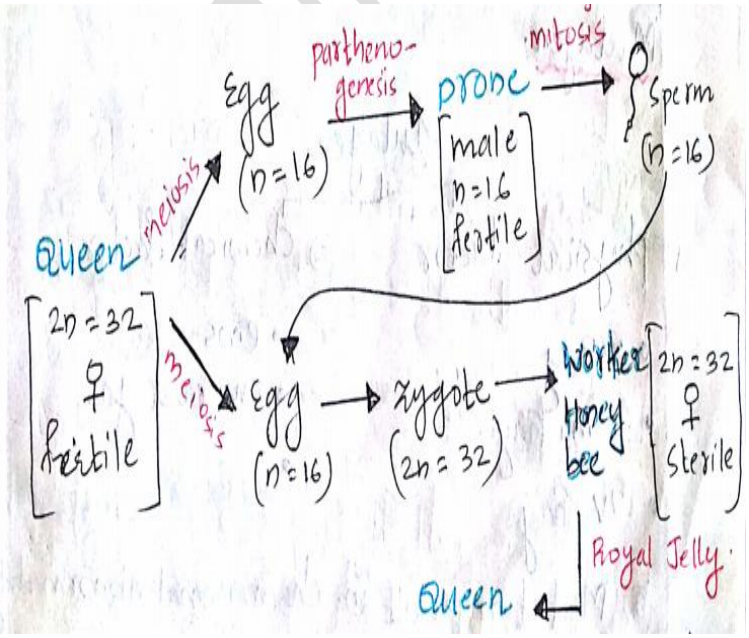
c)XX-XO mechanism-Insects (Grasshopper)

- 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 are heterozygous (XO)and 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



d)Haplo-Diploidy mechanism-Honey bee



- 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.

MUTATION

- Sudden heritable change in the DNA sequences resulting in a change in genotype and phenotype of an organism.
- The substance that cause mutation is called Mutagen

Mutagen

Physical factor	chemical factor
Examples X-ray Gamma-Ray UV-Ray	**Examples** EMS Mustard gas

- Mutation results in chromosomal abnormality (Chromosomal aberrations)
- Chromosomal aberrations are seen in **cancer cells**.
- Mutations are of 2 types
  a)Point mutation
  b)Frame shift mutation

Mutation

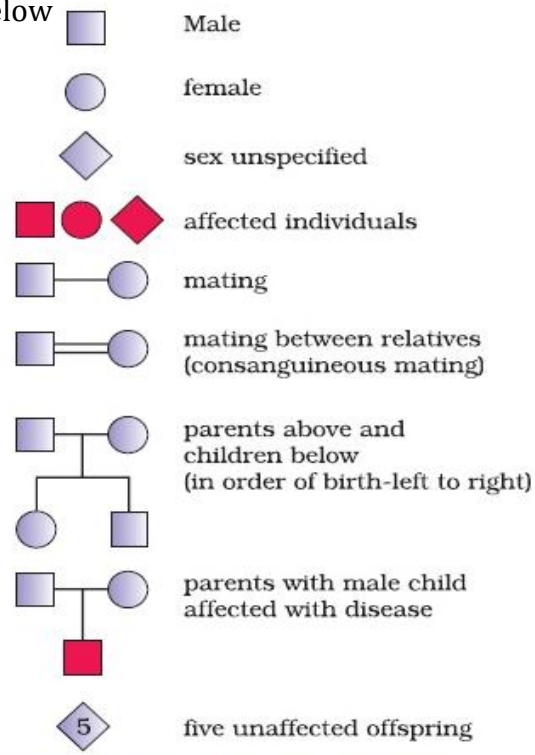
Frame shift mutation	Point mutation
loss(Deletion) or gain (insertion /duplication)of DNA segment results Frame shift mutation. Example: #Thallassemia #cri du chat syndrome	Mutaion due to change in single base pair of DNA Results Point mutation. Example: #sickle cell anemia #PKU

Hugo Devries Proposed mutation theory. He conducted his experiment in Evening prime rose (*Oenothera lamarckiana*)

PEDIGREE ANALYSIS

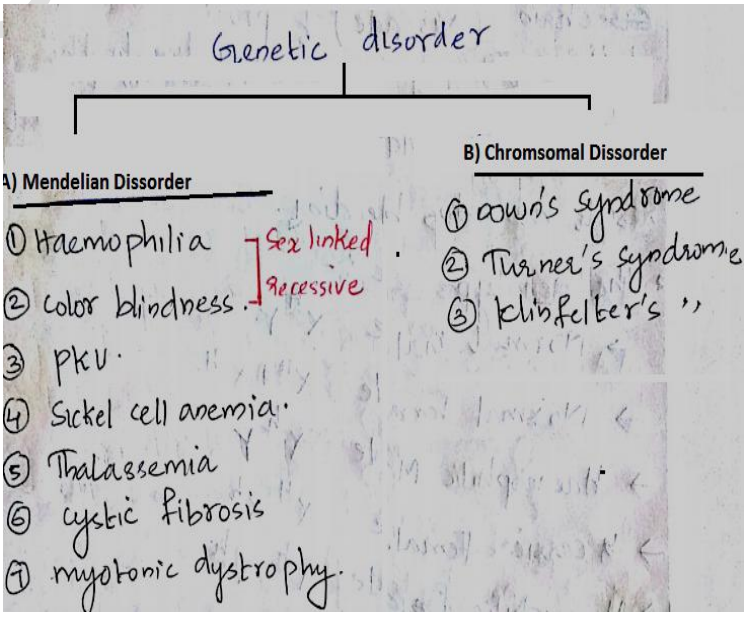
- It is the analysis of trait in a several generations of a family is called pedigree analysis.
- Here inheritance of a particular trait is represented in the family tree (Chart showing family history) over generation.
- This analysis is used to trace the inheritance of a specific trait or abnormality or a disease

- 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.**

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.
- A daughter will not be normally colour blind unless her mother is a carrier and father is a color blind.

3.PKU (PHENYLKETONURIA)

- This is **the autosomal 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

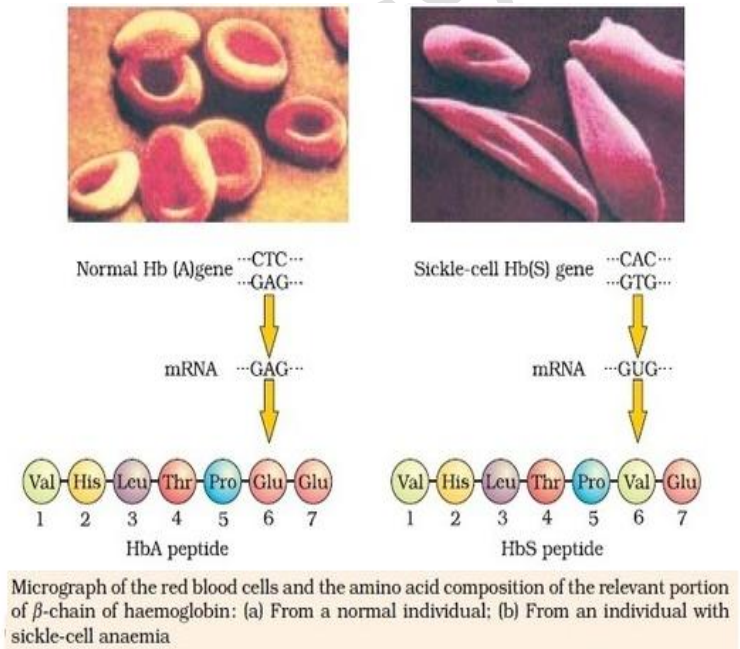
- Normal AA
- Carrier Aa
- Affected aa
- This disease is transmitted from parents to the offspring when both parents are carried (Heterozygous)

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, HbA and HbS .

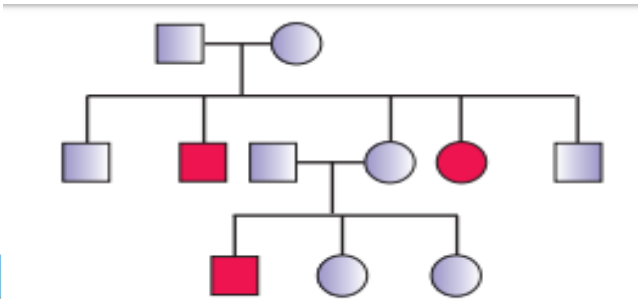
Genotypes are

- Normal $Hb^A Hb^A$
- Carrier Hb^AHb^S
- Affected Hb^sHb^s
- This disease is transmitted from parents to the offspring when both parents are carried (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

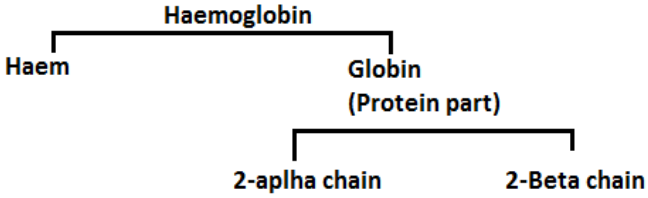


5.THALASSEMIA

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

The genotypes are

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



- Here , production of globin chain is affected. This causes the formation of abnormal Hb resulting in to anaemeia
- 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 genes result alpha thalassemia
- The more gene is affected , less alpha globin molecule is produced

b)β-Thalassemia

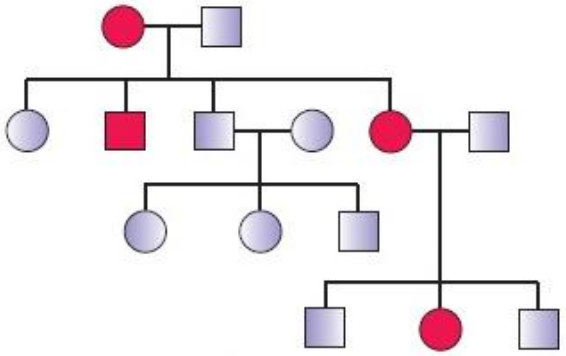
- Here production of Beta globin chain is affected
- α-thalassemia is controlled by a single gene HBB gene.
- this genes are located on the chromosome number 11 of each parent
- Mutation or deletion of one or both genes result Beta thalassemia

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

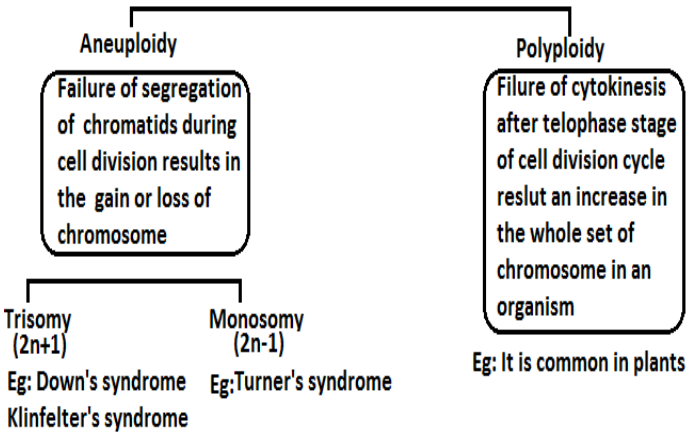
- This is an **autosomal dominant** trait

Pedigree analysis-Myotonic dystrophy



B)chromosomal dissorder

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

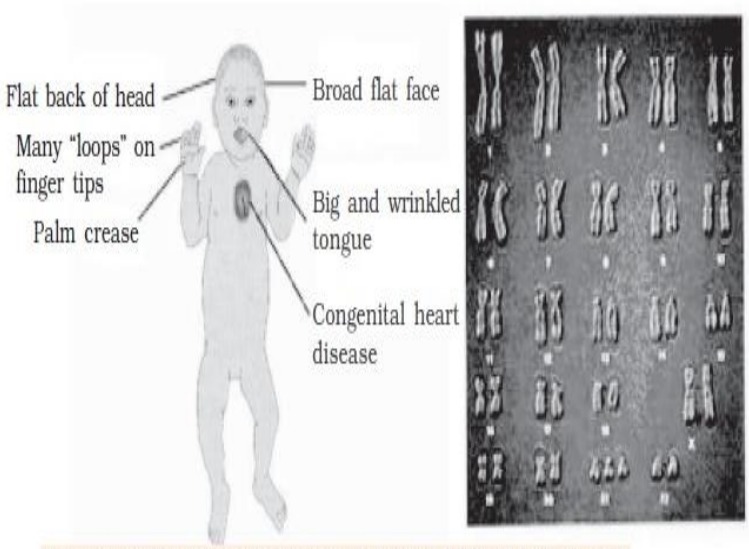


i) Down's Syndrome (45+XX or 45A+XY)

- 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

ii) 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.

- This is due to fetilisation of an abnormal egg (Containing XX) with sperm containing 'Y' chromosome .

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

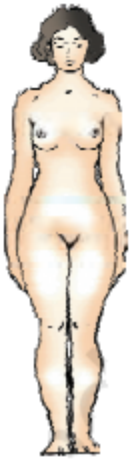
iii) **Turner's Syndrome** : **(44A+XO)**

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

Symptoms

Such females are

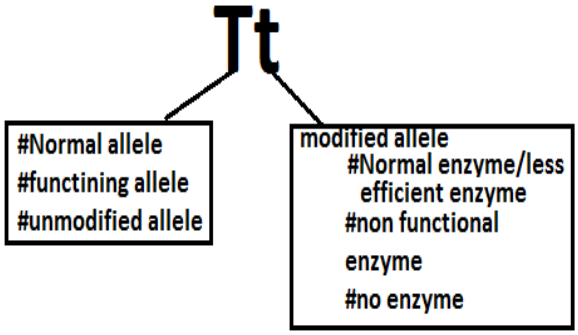
- sterile as ovaries are rudimentary
- lack of other secondary sexual characters



Turner's syndrome

Concept of Dominance

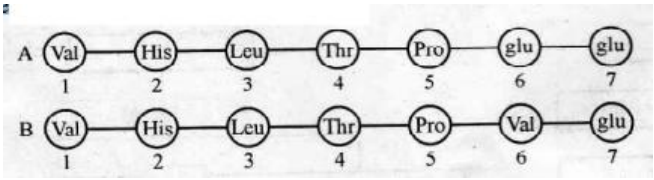
- ✓ In heterozygous conditions (Tt), there are dominant and recessive alleles.
- ✓ The normal allele (functioning allele or unmodified allele)of a gene produce a normal enzyme that is needed for transformation of substrate into product
- ✓ **The modified allele responsible for production of**
  i)Normal 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



Previous year question paper

HSE-March-2019

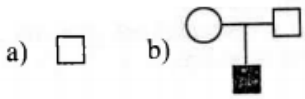
1. "The sex of the baby is determined by the father and not by the mother. Do you agree with this statement? Substantiate your answer (2)
2. Find the odd one out. Justify your answer.
   Down's syndrome, Turner's syndrome, pheny ketonuria, klinfelter's syndrome (2)
3. The amino acid composition of the relevant portion of β chain 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 individual ?

HSE-Model-2019

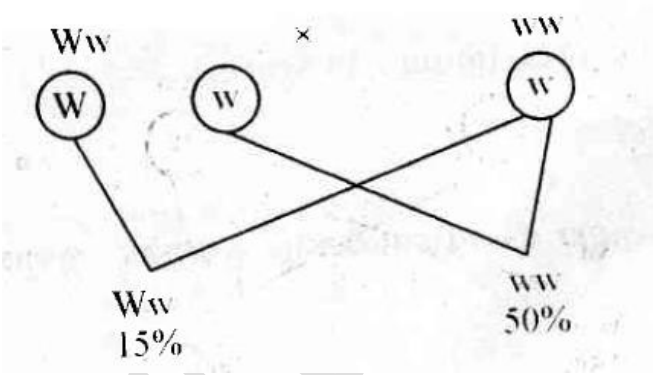
4. crossing of an Fi hybrid into its recessive parent is called (1)
   a) Back cross b) Test cross e) co dominance
   d) Incomplete dominance
5. Drosophila is an ideal material for genetic study. Give 2 reasons. (2)
6. Observe the symbols below



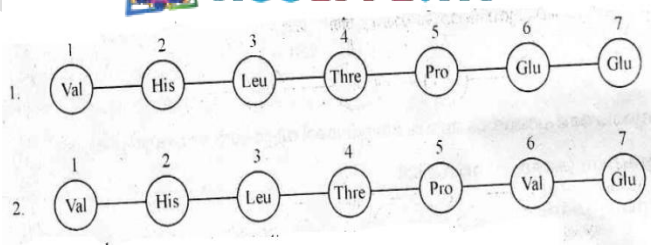
   a) Identify a and b (1)
   b) What is the use of these symbols in genetics? (1)
7. The genetic disorder is caused due to the presence of an additional copy of X Chromosome
   a) Name this disorder
   b) Write the Karyotype of this disorder
   c) Suggest any other characteristic feature of this disorder. (3)

HSE-June-2018

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



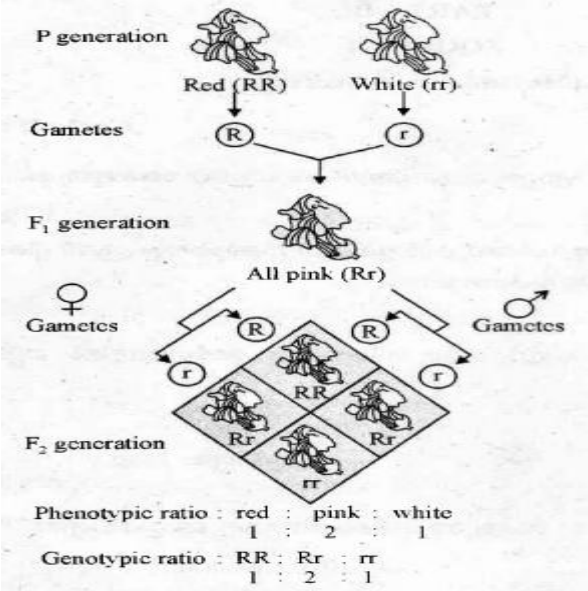
   a) Identify the cross?
   b) Mention the significance of this cross?
9. The following diagram shows amino acid sequences of a part of β chain of haemoglobin of 2 individuals. Observe the amino acid sequence and answer the following questions : (2)



   a)which among the above indicate sickle cell anemic condition ?
   b) justify your answer ?
   c) describe what is single base substitution ?
10. 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

1. 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)
2.



a) Observe the above cross and name this phenomenon?

b) Write down the theoretically given explanation of the phenomenon (2)

3. 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)

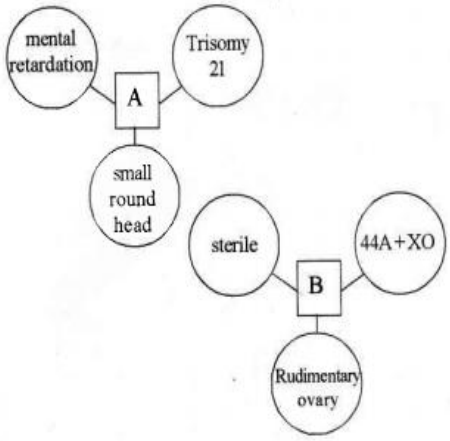
HSE-Model Exam -2018

4. 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)
5. 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?

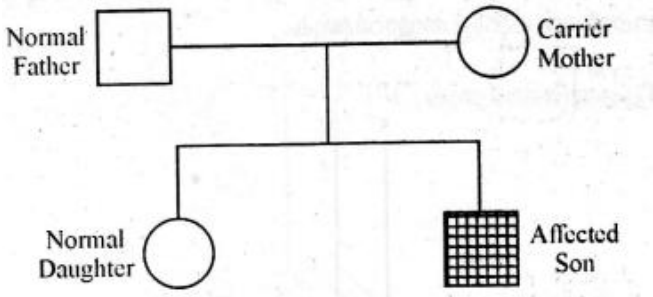
b) Substantiate your answer using co dominance principle. (2)

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



HSE-JUNE-2017

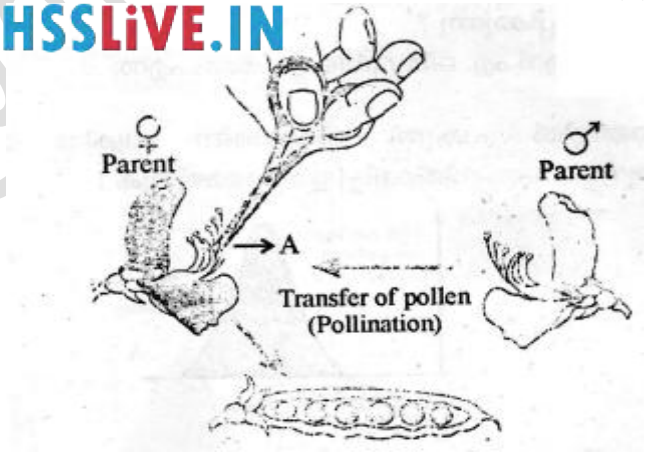
7. Observe the diagrammatic representation of following pedigree analysis and answer the question. (3)



a) Describe the type of inheritance shown in the diagram

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

8. 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

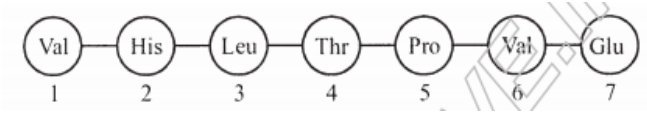
HSE-MARCH-2017

9. 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

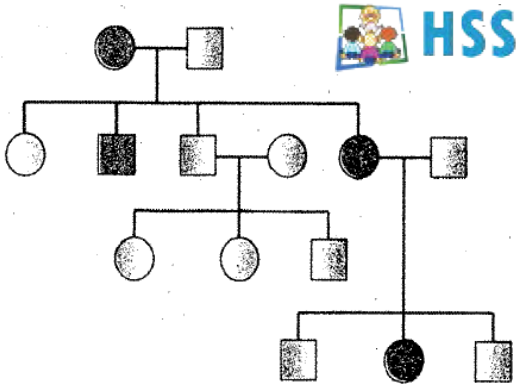
10. Which of the following do not have similar sex chromosome? (homogametic) (1)
    (1) Human female
    (2) Drosophila female
    (3) Bird female
    (4) Bird male

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



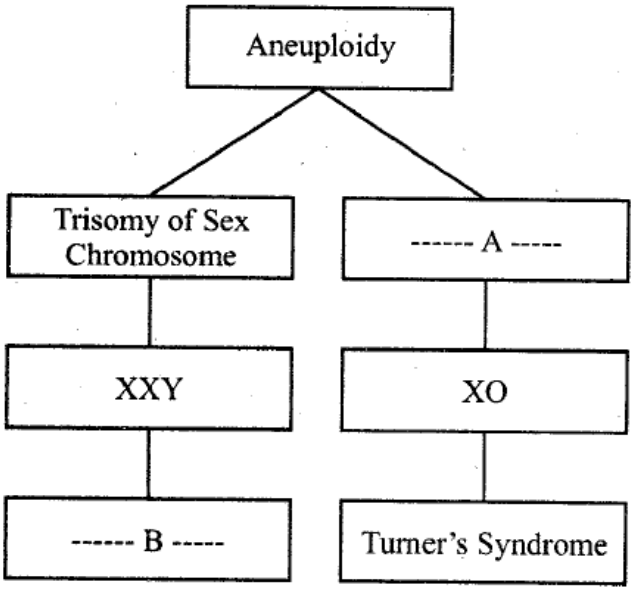
HSE-June-2016

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



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

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

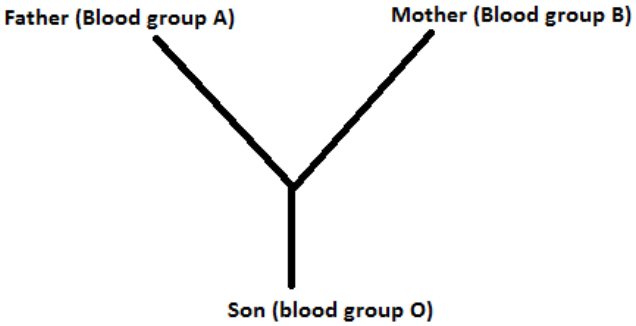


b)What is aneuploidy ?

HSE-March-2016

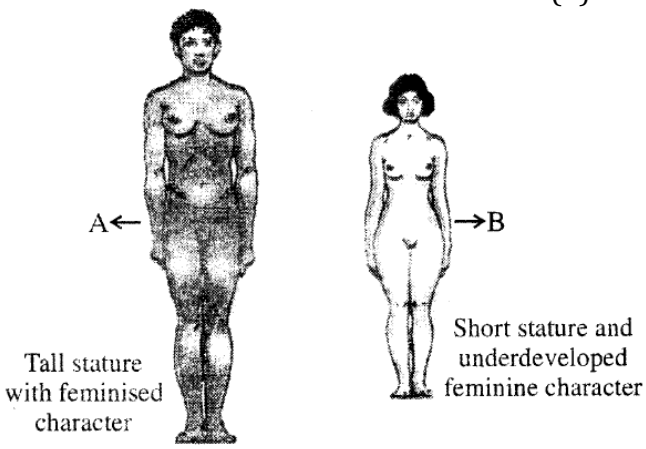
14. Which of the following is not a Mendelian disorder (1)
    Colourblindness, Down's syndrome, Haemophilia, Thalassemia

15. 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)

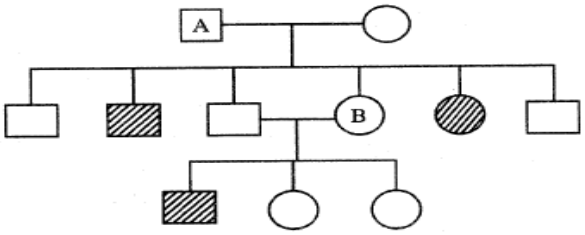
16. Observe the figure and answer the question (2)



a) Identify the syndromes A and B.?
b) What is the chromosome numbers in A and B?

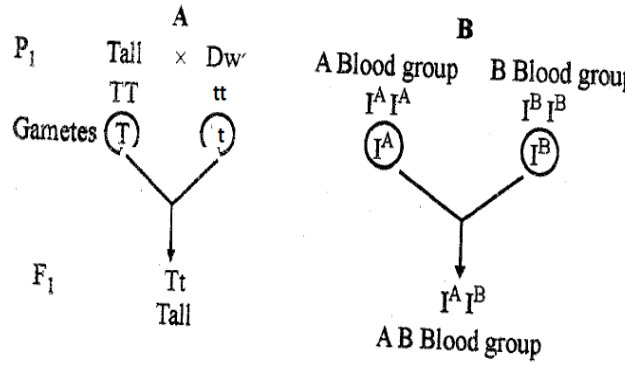
HSE-SAY-2015

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



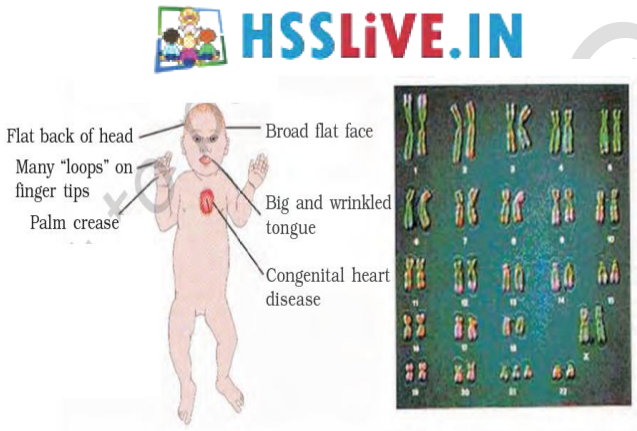
a)Name the type of inheritance shown in the figure ?
b) Write the genotype of A and B?
(Hint : Disease is controlled by a pair of allele Hb^A and Hb^s)
c)Represent pedigree analysis of an X linked Recessive Inheritance diagrammatically

18. 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

19. 

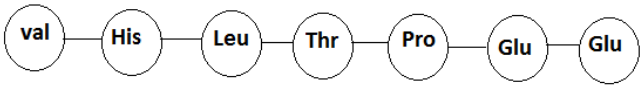
a)Identify the syndrome from the diagram, and write the genotype?
b)It occurs in both sexes (Male and female)? Write the reason (2)

20. 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

21. 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

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



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

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

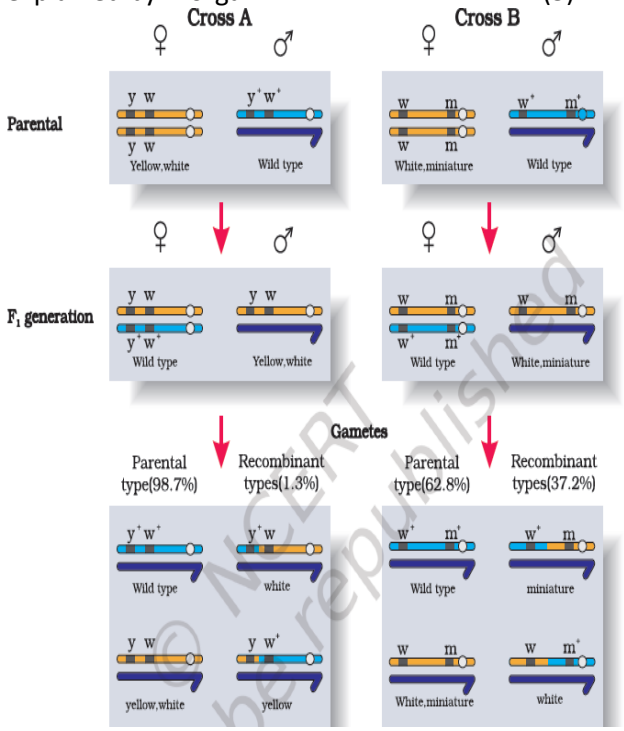
25. a)Define mutation (1)
b)What are the different types of mutation? (1)

26. 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)

27. 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

28. 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)



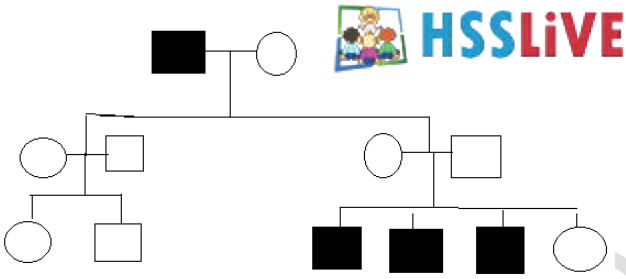
29. Difference in chromosome number of some human being A,B,C, and D is given below:
A)22 pairs of Autosome

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)

30. 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's arguement?
b) Give reason for your argument? (2)

HSE-SAY-2013

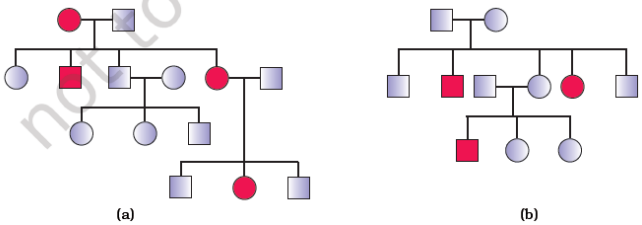
31. In the given pedigree the shaded figure denotes individuals expressing a specific trait (2)



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

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

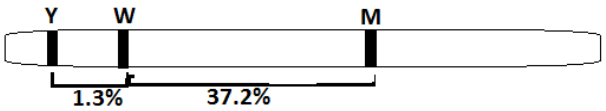


(a) (b)

33. 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

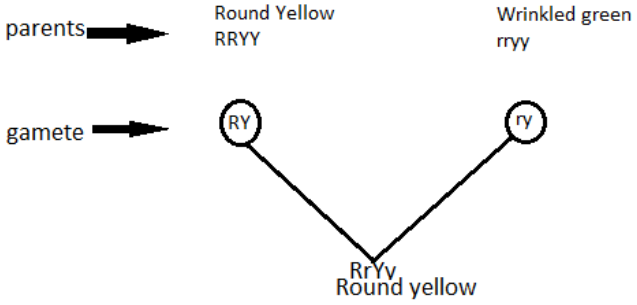
34. Diagrammatic representation of chromosome map of Drosophila is given below (2)



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?

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



a) 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

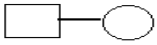
36. Complete the tale using suitable term (2)

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

37. 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)
38. 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

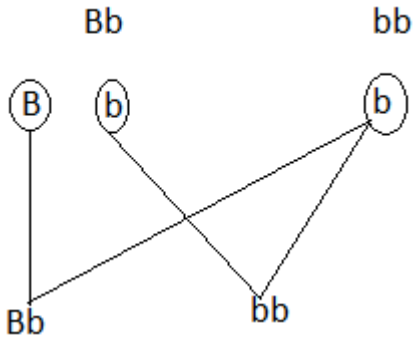
HSE-say-2011

39. 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

40. 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)

41. A genetic cross is represented below (2)



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

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42. 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.

43. 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?

44. 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?

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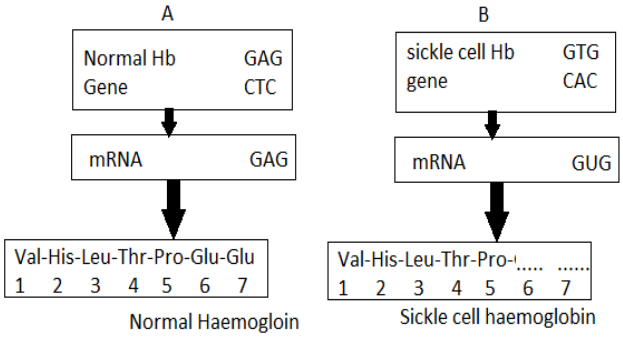
45. Some genetic abnormalities, their genotype and features are distributed in Column A,B and C respectively . Match them correctly (1.5 mark)

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

46. 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

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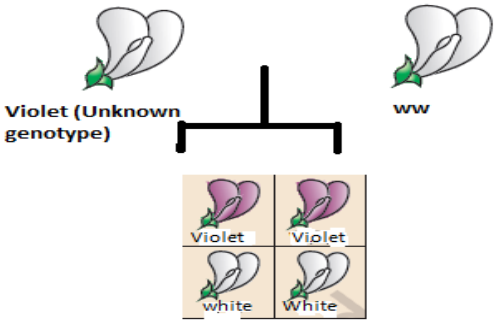
47. To find out the unknown genotype of a violet flowered pea plant a researcher done the flowering cross. Observe the diagram and answer the following question:
(Hint :Violet flower colour in pea plant is dominant over white)



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

48. 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 ?