PRINCIPLES OF INHERITANCE & VARIATION

Genetic Basis of Inheritance

Introduction:

Genetics: It is a branch of biology that deals with the study of heredity and variation.

Heredity: It is transmission of characters from parents to offspring.

Clone: It is product of asexual reproduction, clones are carbon copies of each other derived from a common ancestor (genetically identical).

Variations: It means the differences (Morphological, Physiological, and Anatomical) amongst the individuals of the same species and the offspring of the same parents.

Mendelism:

- Gregor johann Mendel is known as **'Father of Genetics'** Mendel proposed that inheritance is controlled by paired germinal units or factor,Now called gene (by Johannsan).
- Mendel was born in **Silisia**, a village in **Heinzendorf (Austria)** in 1822 to a poor family.
- He passed graduation in 1840 and completed two year diploma in Philosophy.
- Mendel joined on Augustinian monastry of st. Thomas at Brunn (Then in Austria; Now Brno in Czech) in 1843. In 1847, he was made a priest in the monastry.
- Mendel performed hybridization experiment on garden Pea (Pisum -sativum) from 1856 to 1863. After 7 years of experimentation he read out the results of his observations of the Natural history society of Brunn in 1865. **'Natureforschender varien'** was a magazine of german language which was published by **'Natural history society of Brunn'**. In this magazine. The main research paper of **Mendel "Versuche Uber pflanzen Hybriden" or Experiment on plant-hybridization"** was published in 1866.
- Mendel's work remained unnoticed for some **34 years** due to following reasons.
  - **(i)** The scientific world was being rocked at that time by Darwin's theory of evolution (Origin of Species, 1859)
  - **(ii)** Limited circulation of the 'Proceeding of Natural history society of Brunn' in which it was published.
  - **(iii)** Mendel's conclusion about heredity were ahead of his time. Mendel died in 1884 long before his work came to be recognised.

Reason's for Mendel's Success:

Many scientists worked on inheritance. **(Kolreuter work on tobacco, Knight and Goss work on Pea)** But they were not succeed. But Mendel get success because

1. Mendel firstly analyzed the work of former scientists and then prepared his strategy of experiment.
2. Mendel took one or two characters at one time for his breeding experiment.
3. Mendel selected the pea plant for experiment because
   - **(i)** Pea plant is Herbaceous and annual. It can be grown two or three time in a year. So the results obtain very shortly.
   - **(ii)** Pea's flowers are bisexual and self pollinated. So Emasculation and cross pollination can be applied in pea flowers.
   - **(iii)** It can easily grow in garden.
   - **(iv)** Many contrasting characters are found in pea plant. Mendel selected 7 pairs of contrasting traits of pea plant. These characters are as follow.

S.No.	Character	Dominant	Recessive	Chromosome number
1	Plant height	Tall (T) 6 -7	Dwarf (t) 3/4 – 1½	4
2	Shape of pod	Inflated or Full (F)	Constricted (f)	4
3	Position of flower/pod	Axillary (A)	Terminal (a)	4
4	Colour of flower/colour of seed coat	Voilet/Red (V or R) / Grey	White (v or r)/White	1
5	Cotyledon colour	Yellow (Y)	Green (y)	1
6	Pod colour	Green (G)	Yellow (g)	5
7	Seed shape	Round (R)	Wrinkled (r)	7

4. Mendel kept a complete record of every cross and used statistical method and law of probability for analyzing his results.
5. He took care to avoid contamination from foreign pollen grains brought by insects.
6. Mendel selected only pure breeding varieties of pea for his experiment and grew in separate row.
7. Mendel studied the inheritance of character till F_3 generation.

Re-discovery of Mendel's work:

It was in 1900 that three scientists independently rediscovered the principles of heredity already worked out by Mendel. They were **Eric von Tschermak of Austria, Hugo de Vries of Holland and Carl Correns of Germany.**

*** W.Bateson** (1906) firstly used the term **'Genetics'.** He is known as **'father of modern genetics' Muller** established '**Cytogenetics**.

Terminology:

1. **Factor:** Mendelian factor is a unit of inheritance, which passes from one generation to the next through the gamete and controls the expression of a character in the organisms.In modern genetics, It is called **'Gene'** which consists of DNA and genes are responsible for the inheritance and expression of a character.
2. **Allele or Allelomorph:** Contrasting forms of a gene which are found on the same locus in the two homologous chromosomes & control the expression of a trait are called alleles. **Eg: Tallness (T) and Dwarfness (t).**
3. **Phenotype:** It represents the expression of external appearance like colour, shape etc. of an individual.

   Eg: Red colour, Tallness or dwarfness etc.
4. **Genotype:** It indicates the genetic constitution of an individual. **Eg: The genotype of hybrid tall pea plants is Tt, pure tall TT and Dwarf tt.**

- **Johannsan** (1911) firstly used the term Gene, **Phenotype** and **Genotype**.

5. **Homozygous:** It is an individual which contains identical alleles of a gene or factor of a character on its homologous chromosome **Eg: TT and tt**
6. **Heterozygous:** It is an individual which contains the two contrasting forms of a character or two different alleles of a gene on its homologous chromosomes. **Eg: Tt**
7. **Hybrid:** The organism produced after crossing two genetically different individual is called hybrid.
8. **Hybridization:** The process of obtaining hybrid is called hybridization.

9. **Reciprocal cross:** If in one cross individual 'A' is used as male and 'B' as female and in the next cross 'B' is used as male and 'A' as female, It is called as reciprocal cross.
10. **Homologous pair:** Zygotic pair is called Homologous pair.
11. **Back Cross:** It is a cross between F_1 hybrid and one of its parents. Back cross includes test cross.
12. **Test cross:** It is a cross between F_1 hybrid and recessive parent to know whether an individual is homozygous or heterozygous for dominant character.
13. **Genome:** A complete set of chromosomes found in each nucleus of given species is called genome. A single genome is present in haploid cell(n).
14. **Gene Pool:** Sum of all the genes and their alleles present in an interbreeding population is called gene pool.
15. **Phenocopy:** When the different genotypes produce the same phenotypes due to different environments, then one is called the phenocopy of the other **Eg: All seeds irrespective of their genotypes, germinating in dark develop yellow leaves.** Phenocopy is not inheritable. The term **phenocopy** was introduced by Goldschmidt in 1935.
16. **Pure line:** The term was coined by Johannsen in 1900. It is a strain of genetically pure true breeding individuals which have devised by a single self fertilized homozyous ancestor or indentical homozyous ancestors.
17. **Checker Board:** It was firstly used by Reginald C. Punnett (British Geneticist). The analysis of generation is represented in the form of symbols of squares. Female gametes arrange vertically and male gametes arrange horizontally.

Technique of Mendel:

Monohybrid cross:

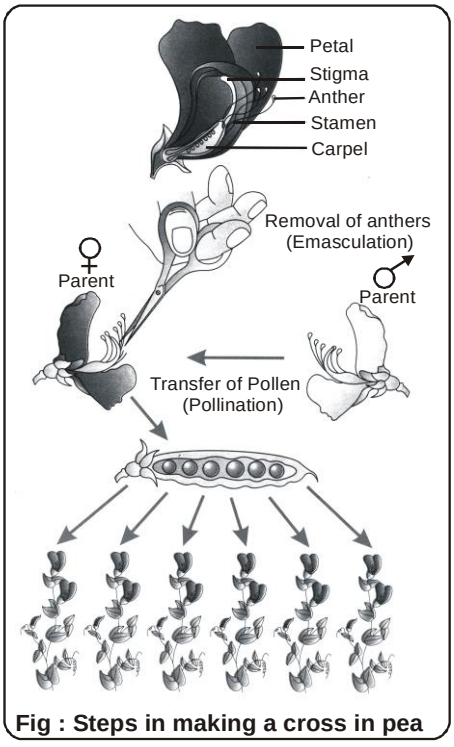
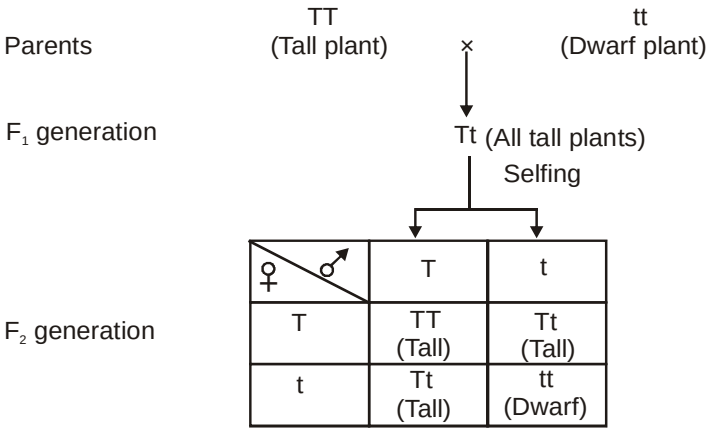


Fig : Steps in making a cross in pea

- It is cross between two organisms of a species that are different in a pair of contrasting traits of a character. **Eg: Height of pea plant.**
- Firstly Mendel selected tall and dwarf plants of garden pea.
- Mendel removed stamen from the flowers of tall plants which were still in bud condition **(Emasculation)**. A bag was then tied over the flowers to prevent cross -pollination **(Bagging).** Dwarf plants were taken as male and their flower was covered with bag.
- On dehiscence or maturity, the pollen grains of dwarf plant were sprayed over the stigma of long plant and It was again covered with bag.
- Seeds were then collected from tall plants. Afterward Mendel obtained F_1 generation through the sowing of former. This process is called **'Hybridization'** and **F_1 generation is called 'Hybrid'.**
- Mendel obtained only tall plants in F_1 generation.
- Mendel applied reciprocal cross but the results were same.
- After this process, Mendel obtained F_2 generation through the sowing of those seeds which were obtained from self pollination of F_1 plants.
- In F_2 generation, Tall and dwarf plants were formed in 3 : 1 ratio and thus it was observed that the character of Tallness in the plants of F_1 generation does not breed pure.
- Mendel used similar technique and procedure for remaining six characters in their experiment and same results came.

The phenotypic ratio of F_2 generation in Monohybrid cross is 3 : 1 and genotypic ratio is 1 : 2 : 1.

❖ Pea plant possess two factors for each (Mendel's choosen) character. The term **'Factor'** was fIrstly used by **Correns.** Which was called **determinor** or **element** by **Mendel**.



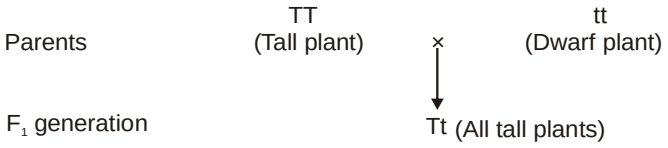
Phenotypic ratio 3 : 1
(Tall) (Dwarf)

Genotypic ratio 1 : 2 : 1
TT Tt tt
(25%) (50%) (25%)

Conclusion of monohybrid cross:

(I) Principle of paired Factors: A plant possesses two factors of each character. Each trait is controlled by a unit factor.

(II) Principle of Dominance: Out of the two factors or alleles representing the alternate form of a trait, one is dominant and expresses it self in the hybrid of F_1 generation. The other factor or allele is recessive and does not show its effect. It is called the 'Mendel's law of dominance'The factor of dominant character is denoted by capital latter and the factor of recessive character is denoted by small letter word.



Law of Dominance

(i) Characters are controlled by discrete units called factors.

(ii) Factors occur in pairs.

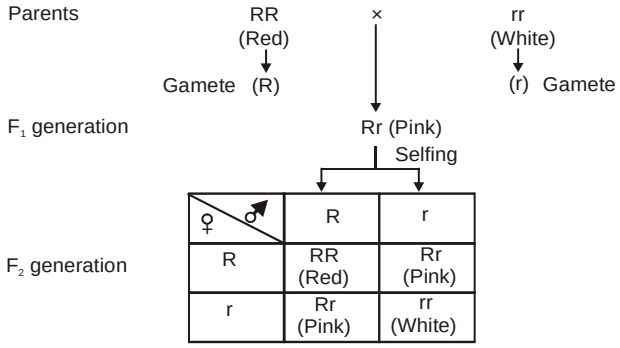
(iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

*** The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F_1 and the expression of both in the F_2. It also explains the proportion of 3:1 obtained at the F_2.**

Exceptions of Principles of Dominance and Paired Factors:

1. Incomplete dominance:

- Incomplete dominance was discovered by **Carl Correns (1903) in *Mirabilis jalapa*.**
  Incomplete dominance is the phenomenon where none of the two contrasting factor or alleles is dominant.
- The expression of the character in a hybrid of F_1 generation is intermediate (partial or mosaic). ***Mirabilis jalapa* is also known as 4 O'clock plant or Gulbansi**
- In Mirabilis jalapa, there are two types of flower colour in pure state, Red and White.When the two types of plant are crossed. The hybrid of F_1 generation have pink flower. If the latter are selfed, the plants of F_2 generation are of three types- Red, Pink and White flowered in the ratio of 1: 2: I
- **The phenotypic and genotypic ratio is similar (1 : 2 : 1) due to incomplete dominance in F_2 generation.**



Phenotypic ratio **1 : 2 : 1**
Red Pink White

Genotypic ratio **1 : 2 : 1**
RR Rr rr
(25%) (50%) (25%)

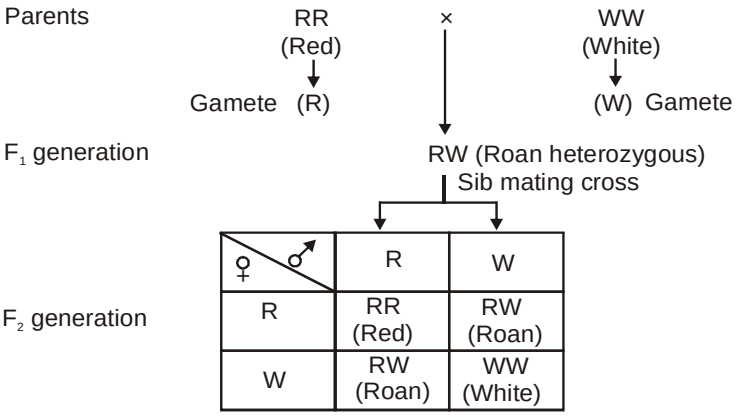
- Incomplete dominance is also found in **colour of flower in Snapdragon or Antirrhinum or Dog flower** and **feather colour in Andulasian fowls**.

Resonate the Concept

- Incomplete dominance is not an example of blending inheritance because the parental types reappear in the F_2 generation. Some worker considered it as example of pattern of quantitative inheritance where single gene pair involved.
  Let's take an example of a gene that contains the information for producing an enzyme. Now there are two copies of this gene, the two allelic forms. Let us assume (as is more common) that the normal allele produces the normal enzyme that is needed for the transformation of a substrate S. Theoretically, the modified allele could be responsible for production of –
  (i) the normal/less efficient enzyme, or
  (ii) a non-functional enzyme, or
  (iii) no enzyme at all
- In the first case, the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait, i.e., result in the transformation of substrate S. Such equivalent allele pairs are very common. But, if the allele produces a non-functional enzyme or no enzyme, the phenotype may be effected. The phenotype/trait will only be dependent on the functioning of the unmodified allele. The unmodified (functioning) allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele. Hence, in the example above the recessive trait is seen due to non-functional enzyme or because no enzyme is produced.

2. Co-dominance:

- In Codominance the alleles are able to express themselves independently in F_1 generation. **Eg: (i) Skin colour in Short Horned cattles**.
- There are two types of pure cattle, Red and White. On cross-breeding the individuals of F_1 generation are found to have Roan colour.Here in a heterozygote of F_1 generation both alleles express themselves independently. The effect is produce due to juxtaposition of small patches of Red and White colour. It is also known as mosaic inheritance. Both the alleles are co-dominant in F_1 generation. On inbreeding the Roan hybrids produce three types of cattle-Red Roan and White in the ratio of 1:2:1.



Phenotypic ratio **1 : 2 : 1**
Red Roan White
1 : 2 : 1
Genotypic ratio **RR RW WW**

Eg : (ii) Sickle cell Haemoglobin. The allele for sickle cell haemoglobin Hb^S is codominant with allele for normal haemoglobin Hb^A.

Eg : (iii) AB Blood Group. Alleles of blood group A (I^A) and blood group B (I^B) are codominant & found over the surface of erythrocytes. When they come together in an individual, they produce blood group AB.

Eg : (iv) MN Blood Group. It is found in humans. The erythrocytes carry two types of native antigens, M and N, and an individual can be MM, MN, NN.

3. Multiple allele:

- **Bernstein (1924) :** Discovered multiple alleles, More than 2 alternative forms of a gene are called as multiple allele. Multiple alleles are located on same locus of homologous chromosome. A diploid individual contains two alleles and gamete contains one allele for a character. **Eg: (i) Human Blood group - 3 alleles**.

Table: Different aspects of blood group

Blood group	Antigen on surface of RBC	Antibody in Plasma	Genotype
A	A	b	I^AI^A, I^AI^O
B	B	a	I^BI^B , I^BI^O
AB	A, B both	Nill	I^AI^B
O	Nill	a, b both	I^OI^O

- Human beings have four blood groups or blood group phenotypes **– A, B, AB and O.**
  A,B,O blood group are determined by allele I^A, allele I^B, allele I° respectively
  I^A = dominant
  I^B= dominant
  I° = recessive
  I^A and I^B are **codominant**.
- If **n** is the number of alleles of a gene then number of different possible genotype = $\frac{n(n+1)}{2}$

  For **eg-** In human blood group number of alleles or **n** are 3. Thus the number of different possible genotype will be **=** $\frac{3(3+1)}{2} = \frac{3\times 4}{2}$ **6 genotypes.**

  Eg: (ii) Coat colour in rabbit

- **Four alleles regulate coat colour in rabbit**

Phenotype	Genotype	Allele type
Wild (Full colour or agouti)	CC', Cc^{ch}, Cc^{h} Cc	C
Chinchilla	$c^{ch}c^{ch}$, $c^{Ch}c^{h}$, $c^{ch}c$	c^{ch}
Himalayan	$C^{h}C^{h}$, $c^{h}c$	C^{h}
Albino	cc	c

Note: These alleles show a gradient in dominance C > c^{ch}> c^{h} > c

Number of possible genotypes = $\frac{4(4+1)}{2} = \frac{4\times 5}{2} = 10$ genotypes.

4. Pleiotropy:

- A gene regulates multiple phenotypic effect. **Eg:** Sickle cell anaemia and Phenylketonuria.

(III) Principle or law of segregation:

- There is mixing of two factor in the hybrid of F_1 generation. At the time of gamete formation in F_1 generation. The two factor separate or segregtate and pass into different gametes randomly. A gamete comes to have one factor of a pair. The gamete fuse randomly during fertilization so that the factors come together in F_2 generation and express themselves freely. So the dwarf plants obtain in F_2 generation.
- Gametes are always pure for a character hence it is also called **'law of purity of gametes'**. **(by Bateson)**

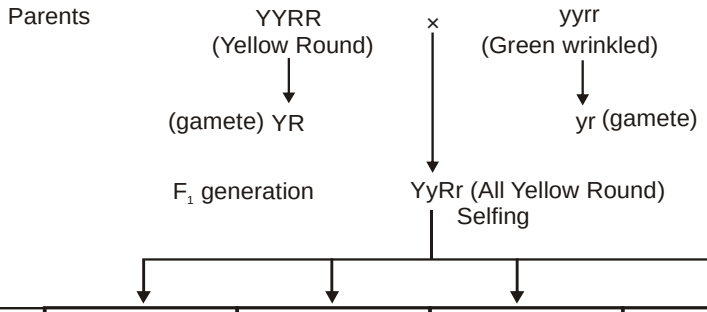
Dihybrid cross:

- It is a cross between two organisms of a species that are different in two pairs of contrasting characters.
- Mendel selected the following two character for this purpose.
  (i) Colour of cotyledon - Yellow and green
  (ii) Shape of seed - Round and wrinkled
- In which Yellow and Round traits are respectively dominant over green and wrinkle traits.
- Mendel performed cross between pure breeding pea plants having yellow round seeds (YYRR) and pure breeding Pea plants having green wrinkled seeds (yyrr).
- All the plants of the F_1 generation were yellow and round seeds (YyRr).

- The factors of both characters will have independently segregated to each other during gamete formation. Thus total four types of gametes (YR), (yR), (Yr), (yr) form in F_1 generation
- On selfing of F_1 the resultant F_2 generation show four types of plants

Yellow Round, Yellow wrinkled, Green Round, Green wrinkled

9/16 3/16 3/16 1/16



F_2 generation

♀ / ♂	YR	yR	Yr	yr
YR	YYRR (Yellow Round)	YyRR (Yellow Round)	YYRr (Yellow Round)	YyRr (Yellow Round)
yR	YyRR (Yellow Round)	yyRR (Green Round)	YyRr (Yellow Round)	yyRr (Green Round)
Yr	YYRr (Yellow Round)	YyRr (Yellow Round)	YYrr (Yellow wrinkled)	Yyrr (Yellow wrinkled)
yr	YyRr (Yellow Round)	yyRr (Green Round)	Yyrr (Yellow wrinkled)	yyrr (Green wrinkled)

Phenotypic ratio **9 : 3 : 3 : 1**

Yellow Round **Yellow wrinkled** **Green Round** **Green wrinkled**

Genotypic ratio **1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1**

YYRR YYRr YyRR YyRr YYrr Yyrr yyRR yyRr yyrr

Thus the **Conclusion of dihybrid cross**

- Number of genotype in any cross is = $\mathbf{3^n}$
- Number of genotypes in dihybrid cross **= 3^2 = 9**
- Yellow round and green wrinkled plants show parental combinations (9+1) while green round and yellow wrinkled show new combinations (3 + 3 = 6). The ratio between parental combination and new combination (recombinants) is 5 : 3: So 62.5% Parental combination and 37.5% new combination (recombinants) are obtained.

Law of Independent assortment:

- Mendel concluded that the two factors of a character assort or separate independent of the factors of other characters at the time of gamete formation and get randomly rearranged in the offsprings. It is called the **'Law of Independent assortment'.**
- The occurence of four types of plants in the F_2 generation of dihybrid cross shows that the inheritance of cotyledon colour assorts independent to the inheritance of seed shape unless green round and yellow wrinkled seeds do not obtain.

Objection:

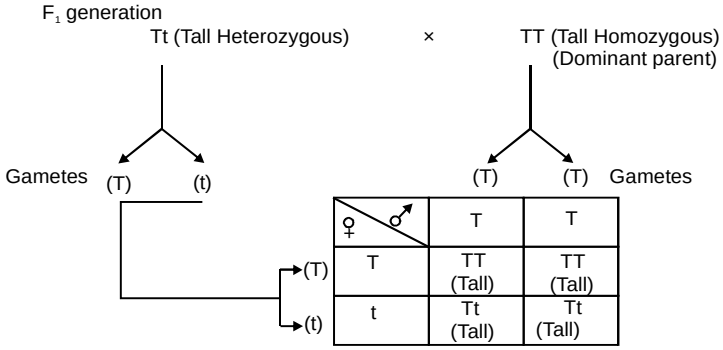
- This law is applicable to only those factors or genes which are either situated distantly on the same chromosome or occur on different chromosomes.

Resonate the Concept

- Occasionally, a single gene product may produce more than one effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b). Starch is synthesised effectively by BB homozygotes and therefore, large starch grains are produced. In contrast, bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds, BB seeds are round and the bb seeds are wrinkled. Heterozygotes produce round seeds, and so B seems to be the dominant allele. But, the starch grains produced are of intermediate size in Bb seeds. So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance. Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.

Back cross:

- It is a cross which is performed between F_1 hybrid and one of its parents.
- If cross is performed between F_1 hybrid and dominant parent then it is called **'Out cross'** All the offsprings obtain from this cross have dominant characters. Thus F_1 generation can be analized through this cross.



(i) Phenotypic ratio = 100% (Tall plants)

(ii) Genotypic ratio = 1 : 1

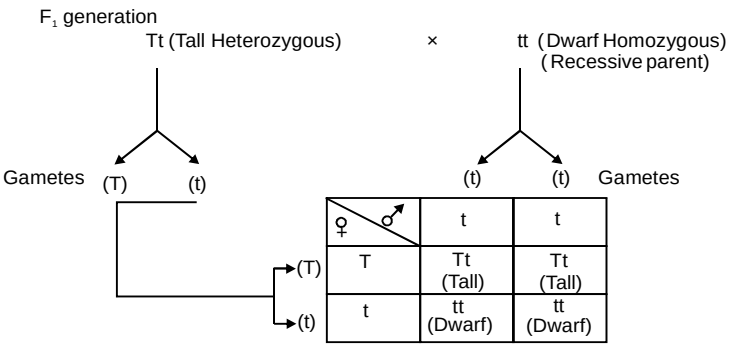
TT Tt

(Homozygous Tall) (Heterozygous Tall)

Test cross:

- Cross is performed between F_1 hybrid and recessive parent. It is a cross to know whether an individual is homozygous or heterozygous for dominant character.

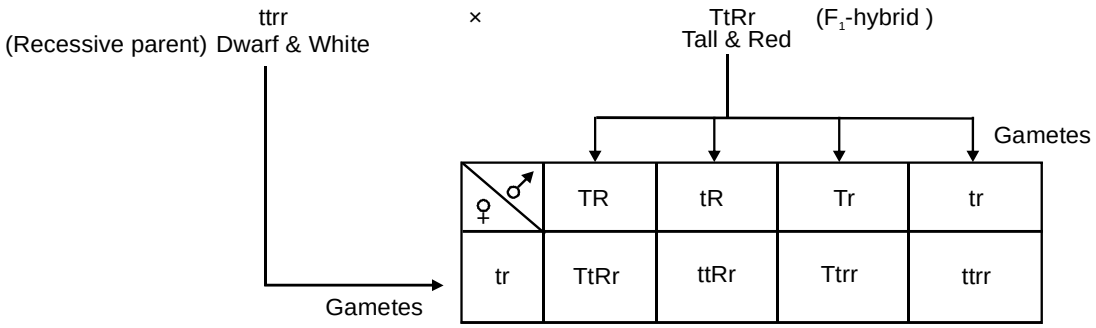
(i) Monohybrid test cross:



(i) Phenotypic ratio 1 : 1
Tall Dwarf

(ii) Genotypic ratio 1 : 1
Tt tt

(ii) Dihybrid test cross:



(i) Phenotypic ratio 1 : 1 : 1 : 1
Tall & Red / Dwarf &Red / Tall & White / Dwarf & White

(ii) Genotypic ratio 1 : 1 : 1 : 1
TtRr / ttRr / Ttrr / ttrr

Importance of Mendelism:

1. It is useful in the field of plant breeding. Advanced features can be used in the development of improved
   varieties of offspring through the hybridization method.
2. The knowledge of dominant and Recessive characters is possible through Mendelism.
3. Development of High productive varieties and disease resistant varieties is possible.
4. It is also useful for the development of improved varieties in animals.

Post Mendelian inheritance:

It involves effect of alleles & non alleles on the normal phenotypic expression of genes. it is of two types
1. Intragenic, 2. Intergenic

1. **Intragenic:** It takes place between two alleles of a gene. eg: Incomplete dominance, Codominance, Multiple alleles. It is interallelic.

   Lethal gene: It was discovered by Cuenot In coat colour of mice. Some genes regulate specific characters in the organisms.

   They cause death of organism if they present in homozygous dominant or homozygous recessive state. If individual dies in embryonic state, it is called Absolute lethality
   Eg 1: coat colour in mice.

Yy × Yy
(Heterozygous yellow) (Heterozygous yellow)

♀ ♂	Y	y
Y	YY Died	Yy Yellow
y	Yy Yellow	yy Brown

Thus Monohybrid phenotypic ratio is modified

2 : 1
Yellow Brown

Yellow body colour (Y) was dominant over normal brown colour (y) Gene of yellow body colour is lethal in homozygous state so that in nature homozygous Yellow mice are never occured in population

Eg 2 : E. Baur discovered lethal gene in Snapdragon (*Antirrhinum majus*).

Cc × Cc
(golden or auria) (golden or auria)

♀ ♂	C	c
C	CC died	Cc golden
c	Cc Golden	cc Green

Phenotypic ratio 2 : 1
Golden Green

Homozygous golden are never occured in nature. If individual dies before reproductive matruity it is called Sublethality Eg: Sickle cell anaemia.

If death of individual takes place after sexual maturity it is called Delayed lethality.

2. Intergenic: It is nonallelic interaction in which two or more independent genes present on same or different chromosomes interact to produce a different expression. It involves following types

(i) Epistasis: In this type of interaction one gene suppresses the expression of another nonallelic gene. The former is called epistatic gene whereas the latter is called hypostatic gene.

Epistasis comprises three types.

(a) Dominant epistasis: In this type dominant gene of one locus suppresses the expression of another gene of different locus.

Eg: Fruit Colour in *Cucurbita pepo* (Summer squash) - A cross between a pure breeding white summer squash (WWYY) with a pure breeding green summer squash wwyy produce white fruits in the F_1 generation.

Dominant gene W is epistatic over Y & w & y genes so that phenotypic ratio of F_2 generation comes to have 12 white fruit, 3 yellow fruit, 1 green fruit.

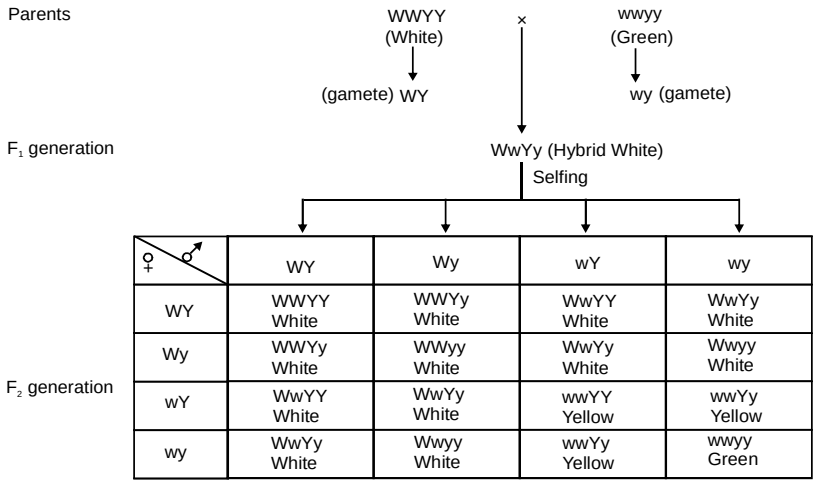


Fig. Dominant epistasis : Inheritance of fruit colour in *Cucurbita pepo*

Phenotypic ratio 12 : 3 : 1
White Yellow green

Other e.g. Coat colour in Dogs (12 white : 3 Black : 1 Brown) and Grain colour in cholam (*Sorghum caudatum*-12 Red : 3 Pearly : 1 Chalky).

(b) Recessive epistasis: In this type recessive homozygous genes of one locus suppresses the expression of another nonallelic gene. Eg: *Coat Colour in Mice*.

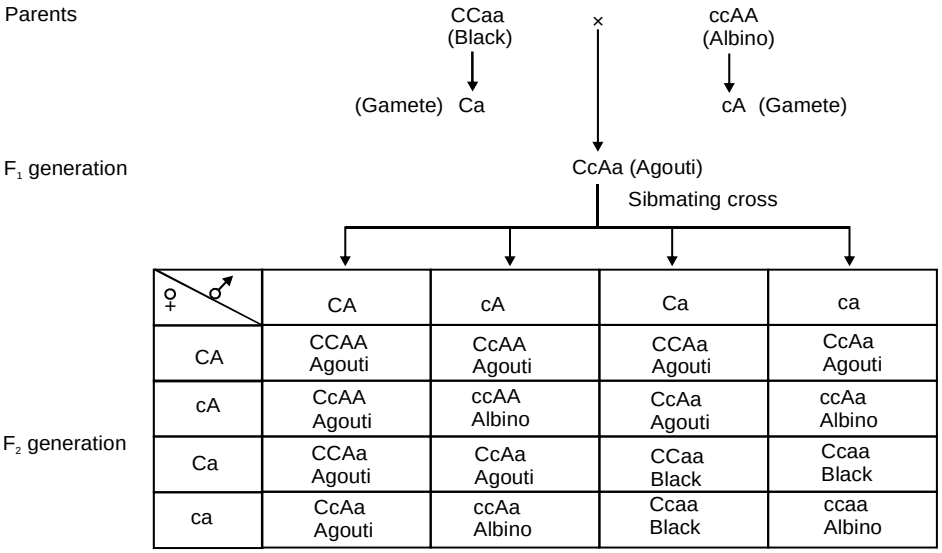


Fig. Recessive epistasis with supplementary gene effect in inheritance of coat colour in mice

Phenotypic ratio - 9 : 3 : 4
Agouti Black Albino

Agouti colour is formed by the occurence of Both C and A gene in either Homozygous or Heterozygous condition. A nonallelic gene c is epistatic in homozygous state (cc) & suppresses the expression of A gene thus albino mice are produced. Other Eg: Pigmentation in Onion Bulb. – 9 Red : 3 yellow : 4 White.

(c) Dominant Recessive Epistasis (Inhibitory gene interaction) : Here the dominant allele at one locus (I) and recessive allele at another locus (cc) give rise to the same effect (I - C, I - cc, iicc). Eg : Plumage Colour in Poultry :

Phenotypic ratio - 13 : 3
White Coloured

In the above cross recessive epistatic gene also produces the same phenotype as the dominant epistatic gene.

(ii) Complementary Genes: In this type. Both nonallelic genes independently produce similar effect when they come together in the dominant form they form a new trait.

Eg: Flower colour of Sweet Pea (*Lathyrus odoratus*) - Flower colour is purple if dominant alleles of two nonallelic genes (C – P –) are present together. The colour of flower is white if the double dominant condition is absent (ccP –, C– pp, ccpp).

Purple colour formation is two step reaction and the two genes cooperate to form the ultimate product.

Raw material A $\xrightarrow{\text{Gene C – Enz}}$ **Chromagen** $\xrightarrow{\text{Gene P – Oxidase}}$ **Anthocyanin**

Parents: CCpp (White) × ccPP (White)

(Gamete) Cp, cP (Gamete)

F_1 generation: CcPp (Purple)

Selfing

F_2 generation

♀ / ♂	CP	Cp	cP	cp
CP	CCPP Purple	CCPp Purple	CcPP Purple	CcPp Purple
Cp	CCPp Purple	CCpp White	CcPp Purple	Ccpp White
cP	CcPP Purple	CcPp Purple	ccPP White	ccPp White
cp	CcPp Purple	Ccpp White	ccPp White	ccpp White

Fig. Complementary gene-Inheritance of purple colour in sweet pea

Phenotyic ratio - 9 : 7
Purple White

(iii) Supplementary Genes: In this type Dominant allele of one gene produces its effect independently where as dominant allele of the second gene is without any independent phenotypic effect but is able to modify the expression of the first gene & produce a new trait. Eg: Seed coat colour in Lablab.

Phenotype ratio - 9 : 3 : 4
Chocolate Khaki Buff

In above example dominant K independently produces Khaki colour. The supplementary gene (L–) changes the expression of pigment forming gene (K) therefore chocolate colour forms.

Other Eg**:** Coat colour in mice - 9 agouti : 3 black : 4 albino

(iv) Duplicate Genes: Two or more independent genes lie on different chromosomes produce same phenotype therefore either they can form the same phenotype in homozygous or heterozygous state.

Independent genes do not form cumulative effect. Eg: Fruit shape in Shepherds's Purse (Capsella bursa).

Phenotype ratio 15 : 1
Triangular Ovoid - Oblong (Top shape)

(v) Polymeric or Additive Genes: In this type two independent dominant genes form same phenotyic effect in homozygous or heterozyous condition. When they come togather then they produce a cumulative effect. Eg: Fruit Shape in Summer Squash (*Cucurbita pepo*).

Phenotype ratio 9 : 6 : 1
Disc Spherical Long

(vi) Collaborative Genes: Two nonallelic genes produce their own phenotypic effect independently when present in the dominant state but can also interact to form a new trait. Eg: Comb types in poultry.

Phenotypic ratio 9 : 3 : 3 : 1
Walnut pea Rose Single

P gene independently produces Pea comb. R gene independently produces rose comb.Both P and R jointly form walnut comb. When none of these genes is present in the dominant state (pprr), single comb is formed.

Table

S.No.	Type of Gene interaction	Phenotypic ratio	e.g.
1.	Dominant Epistasis	12 : 3 : 1	Fruit colour in summer squash
2.	Recessive epistasis	9 : 3 : 4	Mouse coat colour
3.	Complementary gene	9 : 7	Sweet pea flower colour
4.	Polymeric gene	9 : 6 : 1	Fruit shape in summer squash
5.	Duplicate dominant gene	15 : 1	Seed capsule of Shepherd's purse
6.	Collaborative supplementary gene	9 : 3 : 3 : 1	Feather colour of fowl

(vii) Qualitative and Quantitative Inheritance:

(a) Qualitative or Monogenic Inheritance: In this types single dominant gene influences a complete trait. Occurence of two such dominant genes does not alter the phenotype. These genes are called monogenes, **Eg: RR for red colour in Pea.**

(b) Quantitative Inheritance or Polygenic inheritance: If the inheritance of character is controlled by two or more than two genes that is called polygenic inheritance and these

genes are called polygenes or multiple genes in which each dominant allele of a gene produces a part of trait.

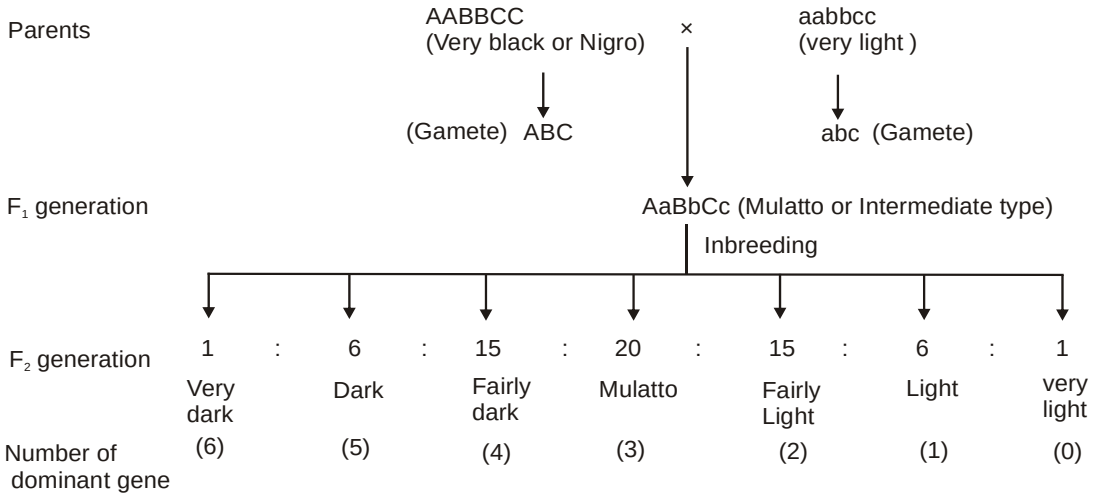
The full expression of trait takes place in the presence of all the dominant alleles of genes. It is called cumulative effect.

It was firstly studied by kolreuter in Tobacco. Francis Galton studied polygenic inheritance of human skin colour.

Nilson Ehle firstly gave the experimental proof of polygenic inheritance. They studied kernal coour of wheat

Eg : 1. Kernal colour of wheat.

Eg : 2. Skin colour of Human (Devenport, 1913)



A Histogram of above cross

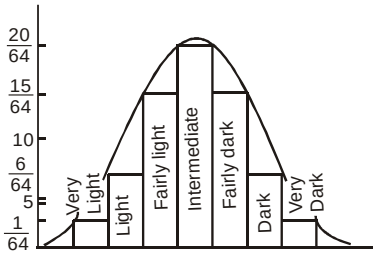


Fig . A histogram prepared from the frequencies of various phenotypes in F_2 generation after a cross between black and white individuals shows a bell shape curve

CHROMOSOMAL BASIS OF INHERITANCE

Chromosomal Theory of Inheritance:

- Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900. Firstly, communication was not easy (as it is now) in those days and his work could not be widely published. Secondly, 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 explaination for the apparently continuous variation seen in nature. Thirdly, Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time. Finally, though Mendel's work suggested that factors (genes) were discrete units, he could not provide any physical proof for the existence of factors or say what they were made of.
- It was proposed by **Sutton and Boveri** independently in **1902** and expanded by **Morgan, Sturtevant and Bridges.**
- Main features of chromosomal theory of inheritance are as follows

(1) Sperm and ovum are bridge between two generations.

(2) Both sperm and ovum contribute equally in heredity of offspring the sperm provides only nuclear part to the zygote.

(3) Nucleus contains chromosomes therefore, chromosomes carry the hereditary characters.

(4) Each chromosome or chromosome pair plays definite role in the development of an individual. Loss of a part / complete chromosome causes structural and functional deficiency in the organism.

(5) Both chromosomes appear in pair in the somatic or diploid cells.

(6) A gamete has only one chromosome as well as one Mendelian factor out of homologous pair.

(7) Synapsis and random independent separation of chromosomes form the quantitative basis of independent assortment of Mendelian factors.

(8) Paired condition of chromosomes is retained during fertilization.

(9) Sex of certain organisms is determined by specific chromosomes called sex chromosomes.

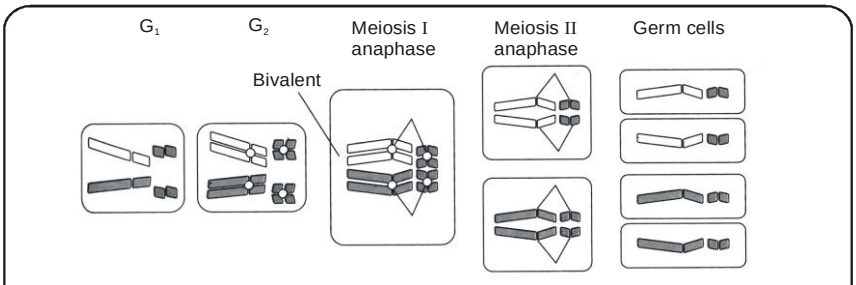
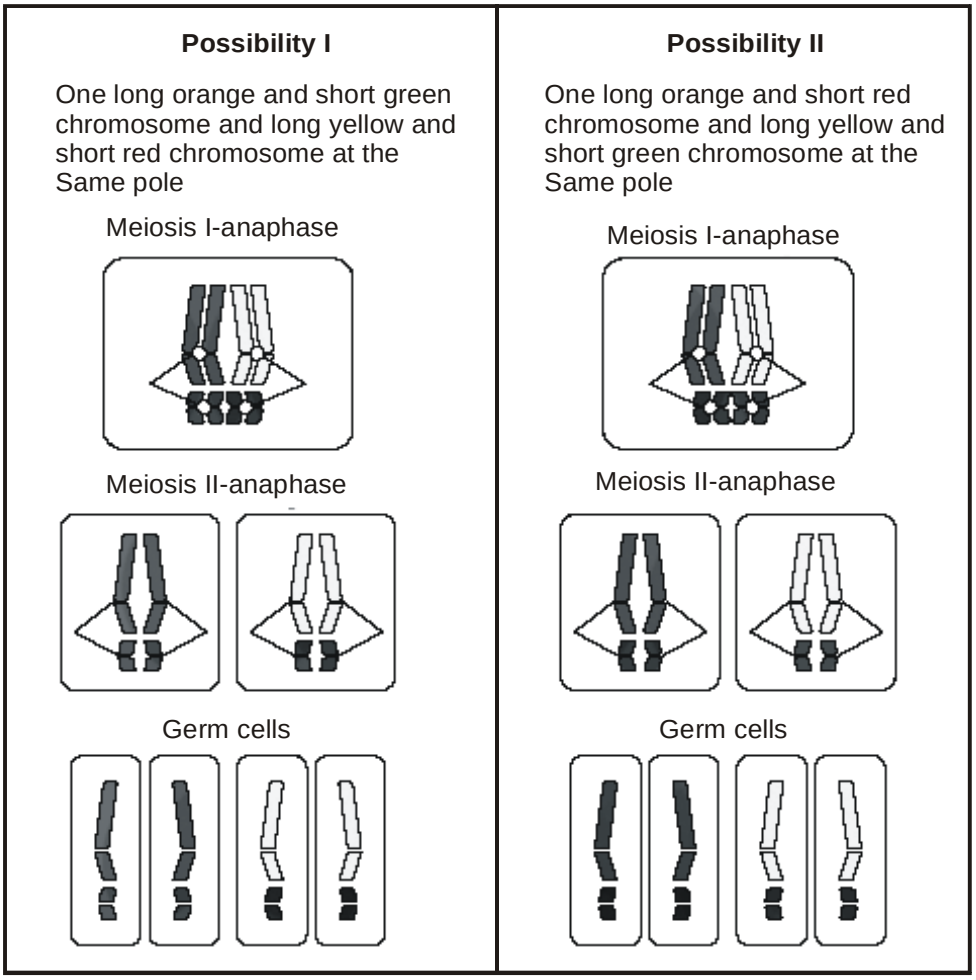


Fig : Meiosis and germ cell formation in a cell with four chromosomes

A Comparison between the behaviour of chromosomes and genes

A	B
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 transmited to a gamete
Independent pairs segregate independently of each other	One pair segregates independently of another pair

During Anaphase of meiosis I, the two chromosome pairs can align at the metaphase plate independently of each other. To understand this, compare the chromosomes of four different colour in the left and right columns. In the left column (Possibility I) orange and green is segregating together. But in the right hand column (Possibility II) the orange chromosome is segregating with the red chromosomes.

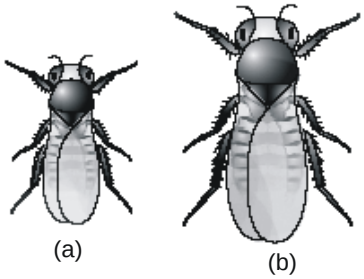


Independent Assortment of Chromosomes

- Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the **chromosomal theory of inheritance**.

  Following this synthesis of ideas, 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. Morgan worked with the tiny fruit files, ***Drosophila melanogaster,*** which were found very suitable for such studies. Because.

  1. They could be grown on simple synthetic medium in the laboratory.
  2. They complete their life cycle in about two weeks,
  3. Single mating could produce a large number of progeny flies.
  4. There was a clear differentiation of the sexes – the male and female flies are easily distinguisable.
  5. It has many types of hereditary variations that can be seen with low power microscopes.



Drosophila melanogaster (a) Male (b) Female

Sex Determination:

- On the basis of fertilization, Sex Determination is of three types.
  - **(i) Progamic:** Sex is determined before fertlization **Eg: drone in honey bee.**
  - **(ii) Syngamic:** Determination of sex takes place during fertilization **Eg: most of the animals and plants**
  - **(iii) Epigamic:** Sex determination takes place after fertilization **Eg: Female in honey bee.**

Usually sex determination involves following types.

(A) Allosomic determination of sex: Chromosomes are of two types.

1. **Autosomes or somatic chromosomes:** They control inheritance of somatic characters.
2. **Sex chromosomes or Allosomes or Heterosomes:** In most of the animals and some plants, sex is determined by sex chromosomes or allosomes. **X-chromosome or X-body** discovered by **Henking** in the **testes of male bug**. **Sex chromosome** discovered by "**Mc Clung**" in **Grasshopper**.

- **Stevens** discovered **Y-chromosome. Wilson and Stevens** (1905) proposed **chromosomal theory of sex determination**. Allosomic determination of sex involves following types.

(i) XX—XY type or Lygaeus type:

- It was firstly described by **Wilson & Stevens in Lygaeus insect.**
- In this types **female** is **homogametic** and forms only one type of gamete whereas **male** is **heterogametic** and produces two types of gametes **Ex: human, Drosophila, dioecious plant like Coccinea, Melandrium.**

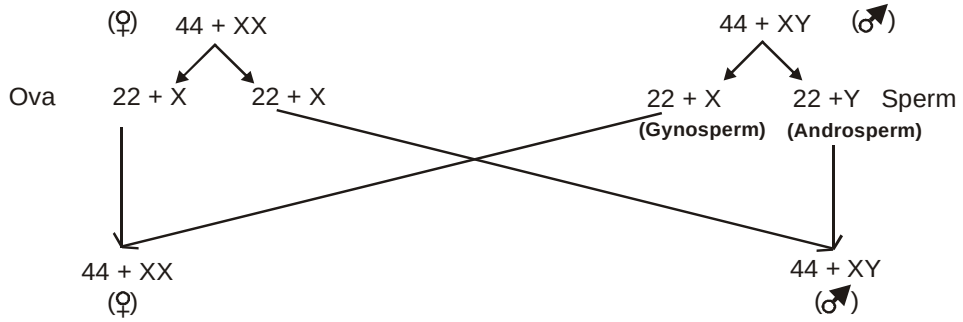


Fig. Sex Determination in human

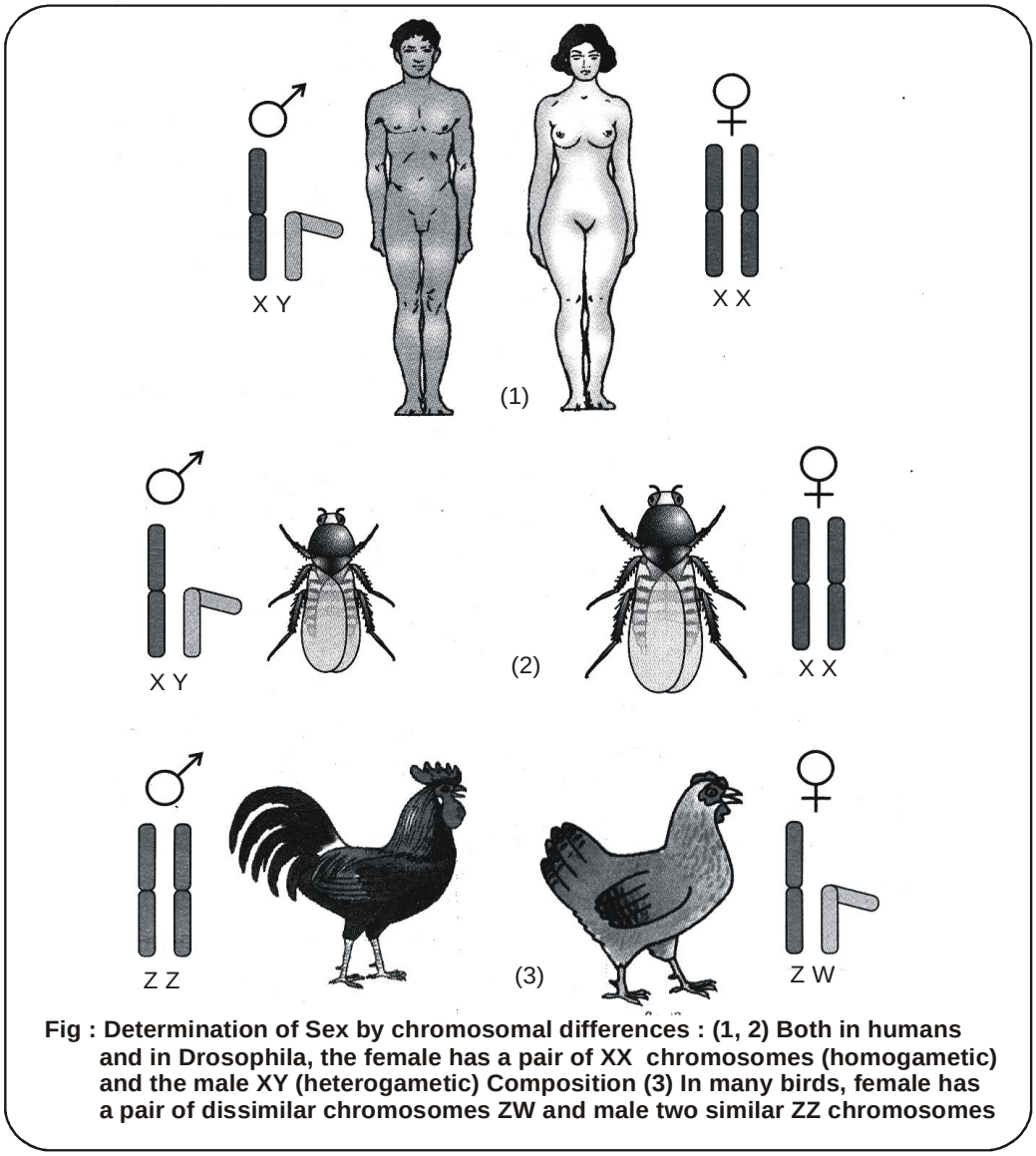


Fig : Determination of Sex by chromosomal differences : (1, 2) Both in humans and in Drosophila, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) Composition (3) In many birds, female has a pair of dissimilar chromosomes ZW and male two similar ZZ chromosomes

(ii) ZW (♀) – ZZ (♂) type:

- Female is heterogametic and produces two types of ova while male is homogametic and produces one type of sperms. **Eg : Birds, Reptiles, Some fishes, Hen, Plant Fragaria eletier.**

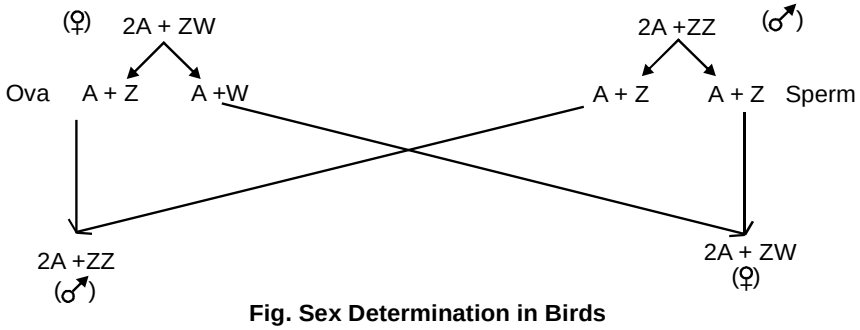
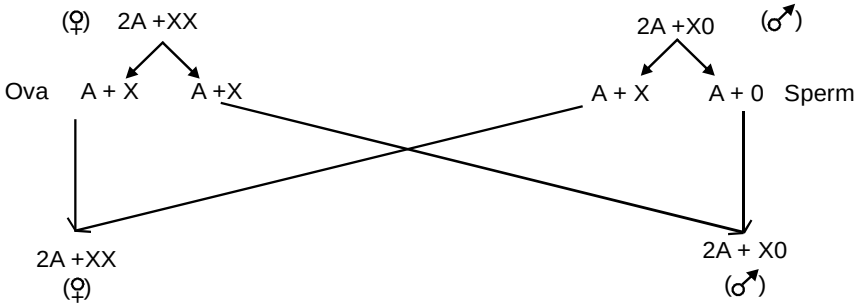


Fig. Sex Determination in Birds

(iii) XX (♀) — X 0 (♂) type or Protenor type:

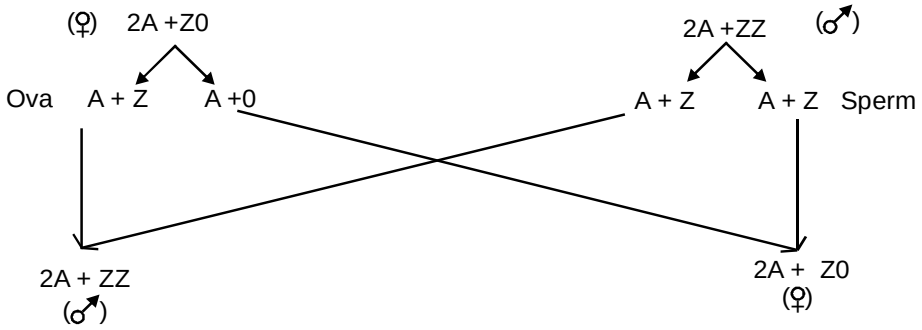
- Female is homogametic and male is heterogametic Actually male bears only one sex chromosome therefore male produces two types of sperm A + X and A+ O. Whereas female produces only one type of egg.

  Eg : Squash bug - Anasa, Grass hopper, Cockroach, Ascaris and plants like *Dioscorea sinuta* & *Vallisneria spiralis*.



(iv) Z0 (♀) – ZZ (♂) type:

- Female is heterogametic in which one sex chromosome is missed Male is homogametic. **Eg: some Moths and Butterflies.**



(v) Haplo-diploidy type:

- In animals such as honey bee female is diploid and male (drone) is haploid.Drone or male insects are develop by unfertilized eggs through parthenogenesis. It is called arrhenotoky. Meiosis does not occur during the formation of sperms. Females develop from fertilized eggs and are hence diploid. Queen Bee takes all the sperms from the drone during nuptial flight and stores them in seminal vesicle. They do not fertilise the eggs & develop into males or drones.

Gynandromorphs:

- Some Drosophila individuals were found to possess half of the body of male and half of female they are called Gynandromorphs. The latter is formed by loss of one X-chromosome at metaphase plate during first zygotic division.

Genic balance theory:

- It was proposed by **Calvin Bridges** (1926). He stated that sex is determined by the genic balance or **ratio** between **X-chromosomes** and **autosomes**. It is applicable for **Drosophila melanogaster** in which **expression of maleness is not controlled by Y-chromosome.**

S. NO.	Chromosome Constitution	X/A ratio	Individual (sex)
(i)	3A + XXX	3/3 =1·0	Triploid female
(ii)	2A + XX	2/2 = 1·0	Diploid female
(iii)	2A + XXY	2/2 = 1·0	Diploid female
(iv)	3A + XX	2/3 = 0· 67	Intersex
(v)	3A + XXY	2/3 = 0· 67	Intersex
(vi)	2A + XY	½ = 0·5	Diploid male
(vii)	2A + XXX	3/2 = 1·5	Super female
(viii)	3A + XY	1/3 0·34	Super male

Linkage:

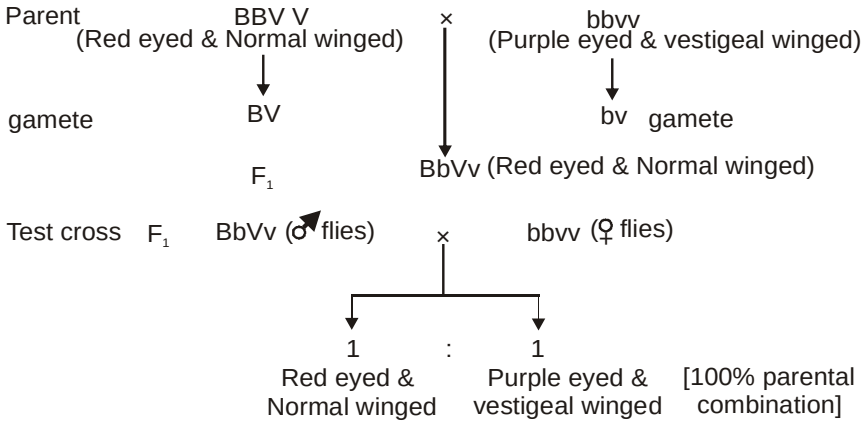
- **Sutten and Bovari** firstly stated about linkage in chromosomal theory of inheritance.
- **Bateson and Punnet** worked on **sweet pea** (***Lathyrus odoratus***) and proposed **coupling and repulsion hypothesis**. They found that the factors for certain characters do not show independent assortment thus **linkage** is **exception of independent assortment**. Therefore the **dihybrid ratio is only 3 : 1 and a test cross ratio of 1 : 1.**
- **According to Morgan (Father of experimental genetics):** The genes of a chromosome have tendency to inherit together and maintains the parental combination in successive generations these genes are called linked genes and this phenomenon is called Linkage.
- **Morgan & casttle** (1911) proposed **chromosomal theory of linkage**
  - **(i)** Linked genes appear in the same chromosome.
  - **(ii)** Genes arrange in Linear fashion on chromosome.
  - **(iii)** The strength of linkage is inversly proportional to the distance between two genes.
  - **(iv)** Genes present on same chromosomes have tendency to mantain parental combinations except for occasional crossovers.

Types of linkage:

(1) Complete Linkage **(2) Incomplete Linkage**

(1) Complete Linkage:

- The gene situated on the same chromosome do not separate and are inherited together over the generations due to the absence of crossing over. It is rare **Ex: Male Drosophila**.



(2) Incomplete Linkage:

- The linked genes do not always stay together because homologous non sister chromatids may exchange segments of varying length with one another during meiosis. It is called crossing over.
- The linked genes located in a chromosome have chances of separation by crossing over, are called incompletely linked genes and the phenomenon of their inheritance is called incomplete linkage. **Eg : Morgan conducted two crosses in Drosophila.**

(i) Crossing of yellow bodied (y) and white eyed (w) female with brown bodied (Y^+) red eyed (W^+) male produced F_1 to be brown bodied red eyed. After intercrossing of F_1 generation Morgan found that F_2 ratio deviated significantly from expected 9 : 3 : 3 : 1 ratio. He observed 98.7% parental types and 1.3% recombinants.

(ii) Second cross performed between white eyed (w w) and miniature winged (m m) female with wild type or red eyed (w^+ w^+) normal winged (m^+ m^+) males. In F_1 generation wild types form. Now F_1 female fly test crossed with white eyed and miniature winged male. He observed 62.8% parental types and 37. 2% recombinants in offsprings.

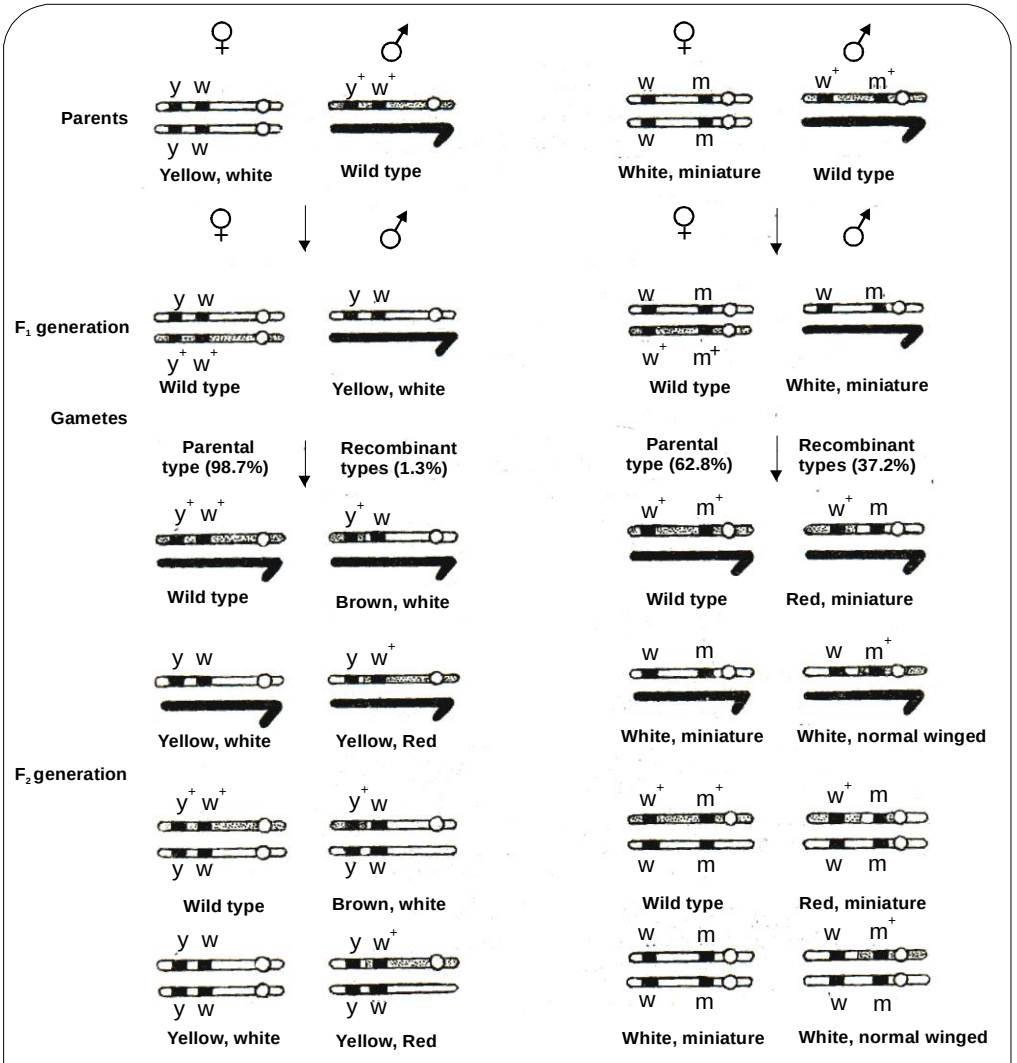


Fig. (i) Dihybrid cross shows limited crossing over between gene y and w . Dominant wild type alleles are represented by (+) sign.

Fig.(ii) Dihybrid cross between white eyed miniature winged female fruitfly with red eyed normal winged (wild type) male fruitfly showing Incomplete linkage.

Linkage Groups:

It is linearly arranged groups of linked genes which are normally inherited together except crossing over or haploid no of homologous chromosomes.

Table

S.No.	Organism	Diploid no. of chromosomes (2n)	no. of linkage group
1	Human beings	46	23
2	Maize	20	10
3	Pea	14	7
4	Drosophila	8	4

* **Neurospora** is haploid. It has **7 linkage groups** (7 chromosomes), **Mucor** has **2 linkage groups** (2 chromosomes).

Sex Linkage or Sex-Linked Inheritance:

❖ Inheritance of some traits is controlled by genes of sex chromosomes. These are called **sex linked traits**. It was discoverd by **Morgan**.

Types of Sex Linked Inheritance: It is of following types.

(i) Criss Cross Inheritance: It is a type of sex linked inheritance where a parent passes the traits to the grand child of the same sex through offspring of the opposite sex.

It is of two types

(a) Diagynic (Diagenic): Father to grandsons through daughter.

(b) Diandric: Mother to grand daughter through son.

(ii) Non Criss Cross Inheritance: It involves two types.

(a) Holandric: Direct from father to son or male to male

(b) Hologynic: Direct from mother to daughter or female to female.

(1) Sex Limited Traits: These traits are expressed in a particular sex although their genes also occur in the other sex they require sex hormones for their expression **Eg: moustaches and beards human males, breast in human females, milk secretion in human females**.

(2) Sex Influenced Traits: These traits are controlled by autosomeal genes & the expression of these genes in a particular sex depends upon favourable conditions. For example the gene of **Pattern baldness** acts as a dominant one in human males because it can express itself due to availability of male hormones.

Genotype	Male	Female
BB	Baldness	Baldness
Bb	Baldness	Baldness Absent
bb	Baldness Absent	Baldness Absent

(3) X-linked genes: In humans X-chromosome carries alleles of a number of human sex-linked disorders like red green colour blindness, haemophilia, glucose-6-phosphate dehydrogenase deficiency syndrome, muscular dystrophy.

(a) Colour blindness:

It is recessive sex-linked trait in which patient is unable to distinguish red or green or blue colours. The gene for the normal vision is dominant. Whereas Colour blindness is recessive to normal vision. **Green colour blindness** is called **deuteronopia**, **red colour blindness** is called **protonopia** whereas **blue colour blindness** is called **Tritonopia** (Rare). Colour blindness can be checked by **ishihara card**.

❖ The normal gene and its recessive allele are carried by X-chromosomes. In man Colour Blindness appears in the presence of a single recessive gene (X^cY) whereas in woman colour blindness occurs only when both the sex chromosomes carry the recessive gene ($X^c X^c$). Colour blindness shows criss-cross inheritance.

Condition -1 female carrier & normal, male Normal.

Parents: XX^c Carrier ♀ × XY Normal ♂

Gametes: XX^c Ova; XY Sperms

Offspring

♀ \ ♂	X	Y
X	XX Normal Girl	XY Normal Boy
X^c	XX^c Carrier Girl	X^c Y Colour Blind Boy

Results : (i) All the Girls are normal (50% normal and carrier whereas 50% normal).
(ii) 50% Boy colour blind and 50% boy have normal vision.

Condition : 2 Carrier female, Colour blind male

Parents: XX^c Carrier ♀ × X^c Y Colour Blind ♂

Gametes: X X^c Ova; X^c Y Sperms

Offspring

♀ \ ♂	X^c	Y
X	XX^c Carrier Girl	XY Normal Boy
X^c	X^cX^c Colour Blind Girl	X^c Y Colour Blind Boy

Results : (i) 50% Girls are Colour blind & 50% normal and carrier Girls.
(ii) 50% Boy colour blind and 50% boy have normal vision.

Condition - 3 Colour blind female, normal male

Parents X^cX^c (Colour Blind ♀) × XY (Normal ♂)

Gametes X^c X^c (Ova) — X Y (Sperms)

Offspring

♀ \ ♂	X	Y
X^c	XX^c Carrier Girl	X^cY Colour Blind Boy
X^c	XX^c Carrier Girl	X^cY Colour Blind Boy

Results: (i) All the Girls are normal and carrier.
(ii) All the Boys are colour blind.

Condition - 4 Normal female, Colour blind male

Parents XX (Normal ♀) × X^cY (Colour Blind ♂)

Gametes X X (Ova) — X^c Y (Sperms)

Offspring

♀ \ ♂	X^c	Y
X	XX^c Carrier Girl	XY Normal Boy
X	XX^c Carrier Girl	XY Normal Boy

Results: (i) All the Girls are normal and carrier.
(ii) All the Boys are normal.
(iii) Occurence of Colour blindness is 0% in the offsprings.

Condition - 5 Colour blind female, Colour blind male.

Parents X^cX^c Colour Blind ♀ × X^cY Colour Blind ♂

Gametes X^c X^c Ova; X^c Y Sperms

Offspring

♀ \ ♂	X^c	Y
X^c	X^cX^c Colour Blind Girl	X^cY Colour Blind Boy
X^c	X^cX^c Colour Blind Girl	X^cY Colour Blind Boy

Results: (i) All Girls & Boys are Colour blind.

FOR AIIMS

(b) Haemophilia (Bleeder's disease) : It was discovered by **John Otto, (1803)** It is Sex linked disease (recessive gene of Haemophilia lies on X- chromosome) in which the patient will continue to bleed even from minor cut . Queen Victoria also infected this disease. Hence it is called Royal disease. It is of two types.

(i) Haemophilia – A: In this type patient does not possess the natural phenomeon of blood clotting due to absence of **factor VIII (AHG or Anti Hemophiliac Globulin).**

(ii) Haemophilia – B: It is due to lack of **factor IX (Plasma thromboplastin factor).**

(iii) Haemophilia – C: It is due to lack of **factor XI (Plasma thromboplastin antecedent)**

It is found in human male only because single gene for the Heamophilia is able to express itself due to presence of one X- chromosome only. This defect does not appear in the girl babies because of the presence of the allele for the normal blood clotting present on the second X -chromosome (XX^h).therefore, the girl babies remain carrier. A female becomes haemophiliac only when both its X-chromosomes carry the gene (X^h X^h). However, such females normally die before birth because the combination of these two recessive alleles is lethal.

Condition -1 female carrier & normal, male Normal

Parents XX^h Carrier ♀ × XY Normal ♂

Gametes X X^h Ova; X Y Sperms

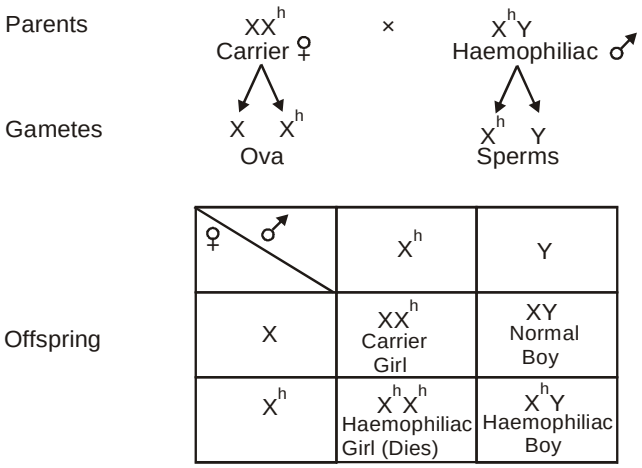
Offspring

♀ \ ♂	X	Y
X	XX Normal Girl	XY Normal Boy
X^h	XX^h Carrier Girl	X^hY Haemophiliac Boy

Results : (i) All the Girls are normal (50% normal and carrier whereas 50% normal)

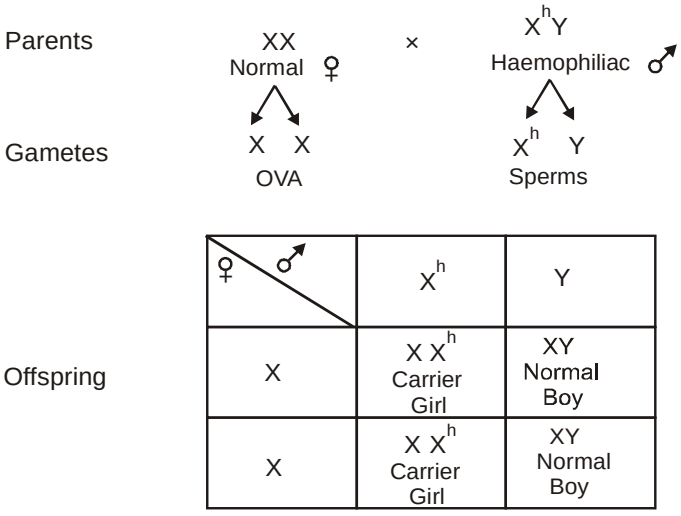
(ii) 50% Boy Haemophilic and 50% boy have normal.

Condition - 2 Carrier female, haemophilic male



Results: (i) 50% Girls are Haemophilic (died) & 50% normal and carrier Girls
(ii) 50% Boy Haemophilic and 50% boy have normal vision.

Condition - 3 Normal female, haemophilic male.



Results : (i) All the Girls are normal and carrier.
(ii) All the Boys are normal.
(iii) Occurence of Haemophilia is 0% in the offsprings.

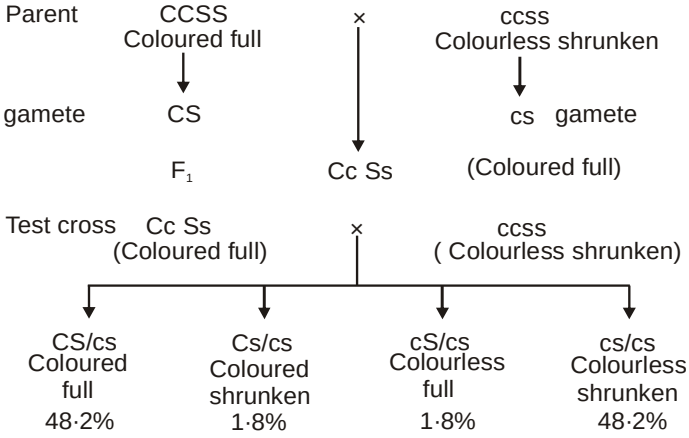
Resonate the Concept

- Homologous part of Y- chromosome (the part that synapses with X-chromosome) bears some similar genes of X-chromosoeme. These are called X-Y linked genes, Eg: Xeroderma pigmentosum (skin cancer), epidermolysis bullosa (blistered skin) in humans.
- **Y- linked gene:** The genes present on diffferential parts of Y-chromosome are passed directly from father to son. These genes are called holandric genes. **Eg: hypertrichosis (excessive hairs on ear pinna), TDF (testis determining factor), keratoderma dissipatum (thickened skin of extremities), porcupine skin, webbed toes in humans, masulatures (mac trait) in male guppy fish (Lebistes).**

Crossing over:

- Exchange of segments of non sister chromatids between homologous chromosomes is called crossing over.
- The term crossing over was coined by **Morgan**. It occurs in the **pachytene stage**, of **meiosis - I**
- The non sister chromatids in which exchange of segments have occurred are called recomobinants or cross-overs while the other chromatids in which crossing over has not taken place are called non cross-overs (parental chromatids).

Eg : Colour & shape of seed / grain in Maize (observed by Hutchinson)



Parental combinations were 48·2% + 48·2 % = 96·4%

New combinations were 1·8% + 1·8% = 3·6%

- In the above cross four types of offsprings obtain in the ratio of 27 : 1 : 1 : 27 instead of 1 : 1 : 1 : 1 ratio expected for independent assortment. Although two genes are linked in the same chromosome but 1.8% nonsister chromatids of the homolgous chromosomes show crossing over.

Factor affecting crossing over:

(i) **Age:** The frequency of crossing over is decreased due to increasing the age.

(ii) **Distance:** If the two genes locate at quite distance. The possibility of crossing over is increased.

(iii) **Temperature:** If temperature increases the possiblity of crossing over also increases.

(iv) **X-rays:** The treatment of X- rays also increases the possiblity of crossing over.

(v) **Heterochromatin:** Occurence of centromere and heterochromatic areas decrease the rate of cross ing over.

(vi) **Chemicals:** The degree of crossing over in animals is changed by chemicals of food.

(vii) **Sex:** Little crossing over is observed in male Drosophila

(viii) **Interference and Coincidence:** One cross over decreases the occurence of frequency of another crossing over. It is called interference. The ratio of observed double cross over and expected double cross over is called coincidence.The latter is small when interference is high.

$$\textbf{Coefficient of coincidence} = \frac{\textbf{\% observed double cross overs}}{\textbf{\% expected double cross overs}}$$

Linkage Maps / Chromosome Maps:

The graphic representation of sequence and relative distance between genes in a chromosome is called Linkage map. The first linkage map was developed by **Sturtevant (1911)** for **Drosophila**. The frequency of crossing over between two genes is directly proportional to the distance between the two.

Recombination frequency or **Cross-over value (COV)** is measured by test cross. 1% crossing over between two linked genes is known as 1 map unit or 1 centimorgan.

$$\textbf{Recombination frequency} = \frac{\textbf{Number of recombinants}}{\textbf{Total number of offsprings}} \times 100$$

or Cross-over value (COV)

Eg: 1 An individual with cd genes crossed with wild type + + On test crossing F_1, the progeny was + c 105, + d 115, cd 880 and + + 900. Distance bewteen cd genes is.

$$\textbf{Recombination frequency} = \frac{\textbf{Number of recombinants}}{\textbf{Total number of offsprings}} \times 100$$

or Cross-over value (COV)

$$\% \text{ cross over value} = \frac{220}{2000} \times 100 = 11 \text{ map unit}$$

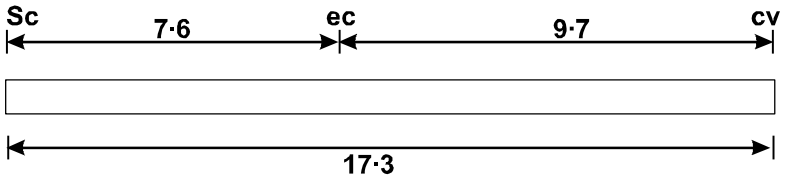
FOR AIIMS

Eg: 2. The frequency of crossing over or recombination of three sex linked genes was found to be

(a) Sc and ec = 7·6% (sc = scute or certain bristles missing)

(b) ec and cv = 9·7% (ec = echinus or rough eyes)

(c) sc and cv = 17·3% (cv = cross veinless or absence of cross veins or wings)



Ans: sequence sc–ec–cv

Mutation:

- Sudden inheritable change in an organism is called mutation.
- **Darwin** coined the term **sports** and **Hugo de vries** coined the term **saltation** for them.
- **Mutation** was discovered by **Hugo de vries** in ***Oenothera lamarckiana*** **(Evening primerose)** He observed 834 mutations in 54343 plants of **Oenothera**.
- Later workers found that 'mutations' observed by De Vries were actually **chromosome aberration and polyploids**.

Types of mutations:

(I) On the basis of direction, mutation involves two types

(1) Forward mutation: Wild type – Mutant type

(2) Backward mutation: Mutant type – wild type.

(II) On the basis of dominance or recessiveness.

(1) Dominant mutation

(2) Recessive mutaiton

(III) On the basis of tissue:

(1) Somatic: It takes place in somatic cell or Vegetative cell. It does not inherit in the next generation but in plants, It can transmit next generation through vegetative propagation.

(2) Germinal: It occurs in germinal cell or reproductive cell. It transmits or inherits from generation to generation.

(IV) On the basis of cytology mutation involves following types.

(A) Chromosomal Mutation (B) Genomatic Mutation (C) Gene Mutation

(A) Chromosomal mutation or chromosmal aberration:

(i) Structural changes & Numerical changes of chromosomes include in this category. Structural changes of chromosomes involve following types.

(a) Intrachromosomal type:

(1) Deletion: A part of chromosome is lost from terminal part (Terminal deletion) or from intercalary part (Intercalary deletion) **Eg: (i) Deletion of a segment of short arm of Vth pair of chromosome in human causes Cri du chat syndrome(cat cry syndrome)** in child **Eg: (ii) Notched wing in Drosophila.**

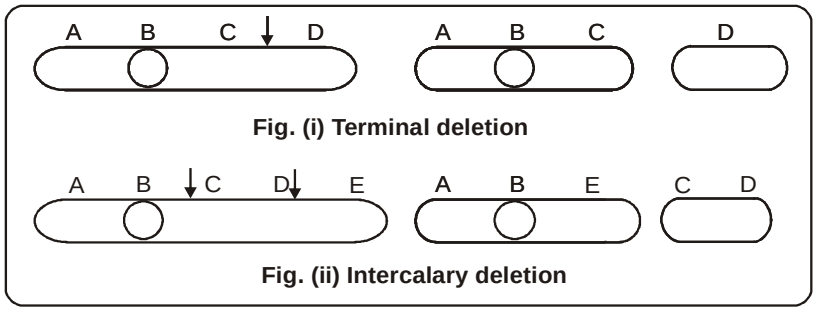


Fig. (i) Terminal deletion

Fig. (ii) Intercalary deletion

(2) Inversion: In this type chromosome break at two intercalary places. The broken segment is inverted up to 180^{o} and rejoined at chromosome. It is called inversion. It is of two types.

(i) Paracentric inversion: Inversion without centromere.

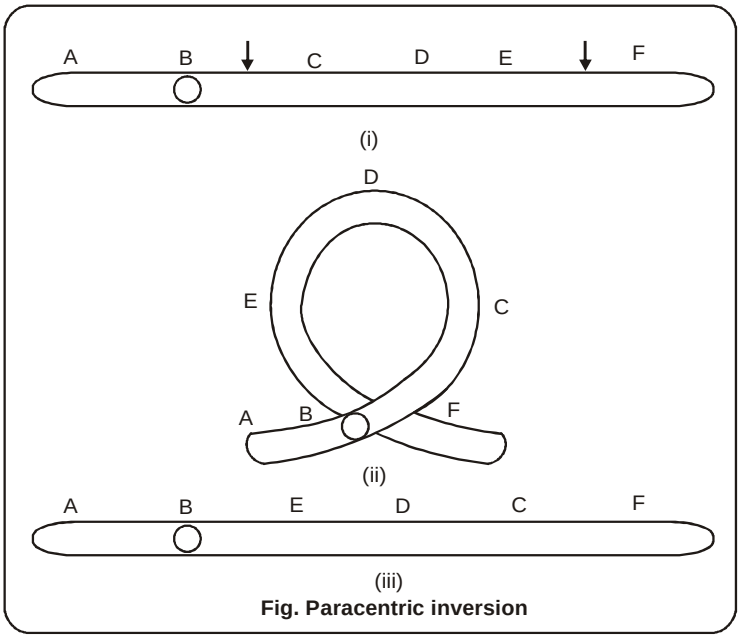


Fig. Paracentric inversion

(ii) Pericentric inversion: Inversion with centromere.

(b) Interchromosomal type:

(1) Duplication:

It occurs due to addition of a part of chromosome.**Eg: Development of Bar eye in Drosophila.** It involves following types.

(i) Tendem duplication: When a chromosomal segement appears two time in a chromosome it is called Tendem duplication.

(ii) Reverse tendem duplication: If the sequence of duplicate part is reverse it is called Reverse tendem duplication.

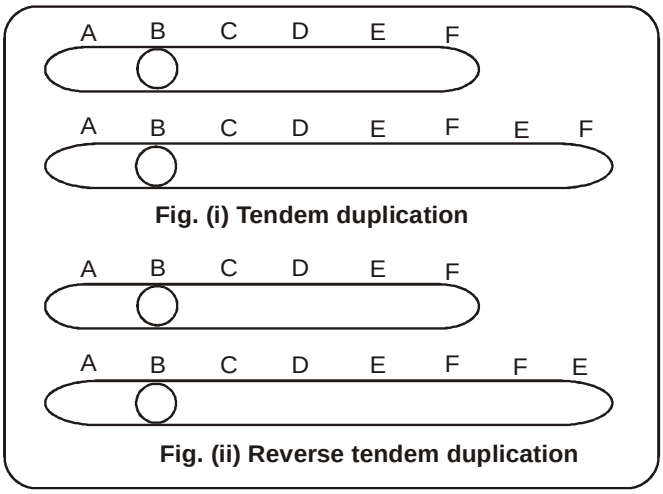


Fig. (i) Tendem duplication

Fig. (ii) Reverse tendem duplication

(2) Translocation:

When the exchange of segments occur between two non homologous chromosomes. It is called translocation.

It is of two types.

(a) Simple translocation: In which a segment of a chromosome transfers to another nonhomologous chromosome.

(b) Reciprocal translocation: In which a mutual exchange of chromosomal segments between two non homologous chromosomes. **Eg: Chronic myeloid leukemia** is due to reciprocal translocation in between 9th and 22nd chromosomes.

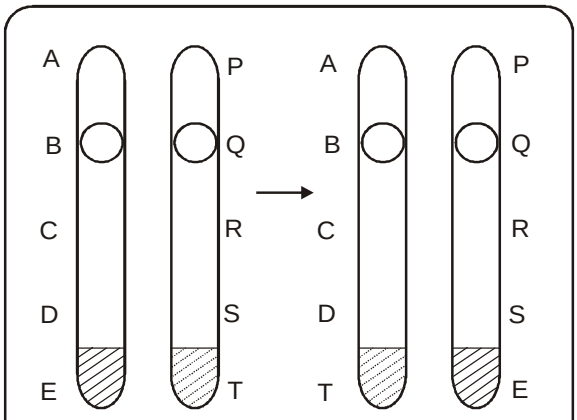


Fig. Reciprocal translocation

(B) Genomatic Mutation: (Changes in chromosomal Number) It involves **aneuploidy and Euploidy (polyploidy).**

(1) Aneuploidy: In which one or few chromosomes are either deficient or in excess in a species. It is of two types.

(a) Hypoploidy: In which either one or few chromosomes are deficient. It involves two types.

(i) Monosomic (2 n – 1): In which one chromosome is deficient. **Eg: Turner's syndrome (44 + X 0),** It is also found in Gossypium.

(ii) Nullisomic (2n – 2): A pair of chromosome is deficient.

(b) Hyperploidy: In which one or few chromosomes are in excess. It is of two types.

(i) Trisomy (2n + 1) : One chromosome is additional in a pair of chromosome. It represents as 2 n + 1. **Eg : Datura,** In human Trisomy in 21^{st} pair,Trisomy in 18^{th} pair and Trisomy in 13^{th} pair are respectively called down's syndrome. Edward syndrome and patau syndrome. **Double trisomic** has two different chromosomes in triplicate **(2n + 1 + 1)**.

(ii) Tetrasomic (2n + 2): A pair of chromosome is additional or a chromosome is found fourtimes **Eg : Super female 44 + XXXX, Double tetrasomic (2n + 2 + 2).**

(iii) Pentasomic (2n + 3): A chromosome occurs five time **Eg : Superfemale (44 + XXXXX) rare.**

Resonate the Concept

- **Double monosomic:** In double monosomic one chromosome is deficient in each chromosome of a pair. It represents as **2n – 1 – 1**.
- **Mixed Aneuploids:** In which both hypoploidy and hyperploidy occur in two different pair of chromosome. **Eg: 2n + 1 A – 1B**.
- **Euploidy:** In which chromosome number is exact multiple of genome **Eg: Monploidy, diploidy, polyploidy.**

Polyploidy:

- If an organism has more than two sets of chromosomes, It is called Polyploid this phenomenon is called polyploidy.
- On the basis of occurence of number of genome, polyploid is called **triploid (3n), tetraploid (4n), Pentaplaoid (5n), Hexaploid (6n)** etc**.**
- In nature, polyploidy appears due to the failure of chromosomes to separate at the time of **anaphase** either due to **nondisuction** or due to **noformation of spindle**.
- Polyploids with odd number of genomes **(Eg: triploids, pentaploids)** are sexually sterile. So that they perform reproduction by vegetative propagation **Eg: Banana, Pineapple**.

Polyploidy involves three types:

(i) Autopolyploidy **(ii) Allopolyploidy** **(iii) Autoallopolyploidy**

(i) Autopolyploidy:

- It is a numerical increase of the same genome such as Autotriploidy (AAA) **Eg : Rice, Gram, Maize**.

(ii) Allopolyploidy:

- It is formed by hybridisation between two species followed by doubling of chromosomes such as (AABB) **Eg : Wheat, Tobacco, artificially produced two allopolyploids are Raphanobrassica and Triticale.**

(iii) Autoallopolyploidy:

- One genome is in more than diploid state such as (AAAABB) **Eg: *Helianthus tuberosus***.

Resonate the Concept

- **Polyploidy can be artificially induced by application of colchicine and granosan**.

(C) Gene Mutations:

- Sudden stable changes in the structure of gene or cistron due to change in nucleotide sequence and nucleotide type are called gene Mutations.
- Most of the gene mutations include change in single nucleotide. These are called **point mutations.** If mutations takes place in more than one base pair is called **gross mutation**. Usually Gene mutations appear during replication of DNA therefore it is called **copy error mutation.**
- **Seth Wright** (1791) firstly recorded point mutation. He observed **short legged lamb (ancon sheep).**
- Scientifically **Thomas Hunt Morgan** observed actual mutation (gene mutation) when he found white eyed mutant of male Drosophila among wild type red eyed Drosophila.

Type of gene mutations: Gene mutations involve three types

(i) Substitution: It includes replacement of one type of nitrogenonus base by other. It is of two types

(a) Transition: In this type, one purine is replaced by another purine while one pyrimidine by another pyrimidine.

(b) Transversion: Purine base is replaced by a pyrimidine base or vice versa.

(ii) Inversion: A deterioration of DNA by mutagen can change the base sequence of a cistron in the reverse order.

(iii) Frame-shift Mutations: In this types the entire reading frame of base sequence shifts laterally either in the forward direction due to insertion of one or more nucleotides or in the backward

direction due to deletion of one or more nucleotides. It is also called **gibberish mutations.** It is of two types.

1. **Deletion:** One more nucleotides are eliminated from segment of DNA or gene.
2. **Insertion:** Addition of one or more nucleotides in the segment of DNA or gene.

On the basis of occurrence in nature, mutations two types.

1. **Spontaneous Mutations:** They occur naturally in the nature due to internal factors such as
   (i) Background Radiations
   (ii) Tautomers.
   (iii) Copy Error
   (iv) Deamination of Cytosine

2. **Induced Mutations:** they are produced by the application of external factors and chemicals. The first induced mutation produced by **Muller (1927)** in **Drosophila** through the use of **X-rays**.

   Mutagens: Any type of physical or chemical factor that is used in the induction of mutation are called mutagens. The latter are of two types.

   (i) Physical mutagens: These are of two types
      (a) High Energy Radiations: These are of two types
         (1) Ionizing radiations: Eg: X-rays, gamma rays and cosmic rays, α–rays, β– rays. Ionizing radiations cause breaks in the chromosome. It causes abnormal cell divsions. Different types of cancers are the result of radiations.
         (2) Non-ionizing radiations: Eg: Ultra-violet light. Thymine (pyrimidine) dimers show a major mutagenic effect of UV rays that disturbs DNA double helix and thus DNA replication.
   (ii) Chemical Mutagens: They are of several types.
      (a) Base Analogues: They resemble the normal bases of DNA and, therefore, get incorporated into DNA in place of them. The common mutagens of this type are **5-bromouracil and 5-flurouracil**.
      (b) Alkylating Agents: Nitrogen mustards **Eg: RN (CH_2Cl_2) diethyl sulphate (DES), dimethyl nitrosamine (DMN) and other alkylating agents** cause methylation or ethylation of nitrogen bases.
      (c) Nitrous Acid: It is a deaminating agent which changes cytosine to uracil, guanine to xanthine and adenine to hypoxanthine. It disturbs replication and transcription.
      (d) Acridines: They are derived heteroaromatic flat molecules from which a number of dyes and pharmaceuticals are prepared. **Eg: Acriflavine, proflavine, acridine orange. Deletion or addition of nucleotides cause frame shift mutation.**

Resonate the Concept

- **Same-sense mutation or silent mutation:** In this type the codon is changed but the It does not alter the amino acid specificity **Eg : (CCA CCC , GCC GCG)**
- **Mis-sense mutation:** When the meaning of codon changes from one amino acid to another it is called missense mutation **Eg : Human disorder called sickle cell anaemia is caused by a single base substitution in a gene**. This alteration results in the replacement of **glutamic acid** by **valine**.
- **Non sense mutation:** In this types polypeptide synthesis is stopped due to formation of a terminating or nonsense codon, such as **UAA, UAG, UGA.**

Human Genetics:

- **Archibald Garrod,** firsty studied human genetics. He studied black urine disease alkeptonuria and stated that it is inherited by recessive gene. This was also the first study of inborn error of metabolism and biochemical genetics, hence Garrod is also known as **"Father of human genetics" or "Father of biochemical genetics".**

Important techniques used to study human genetics are.

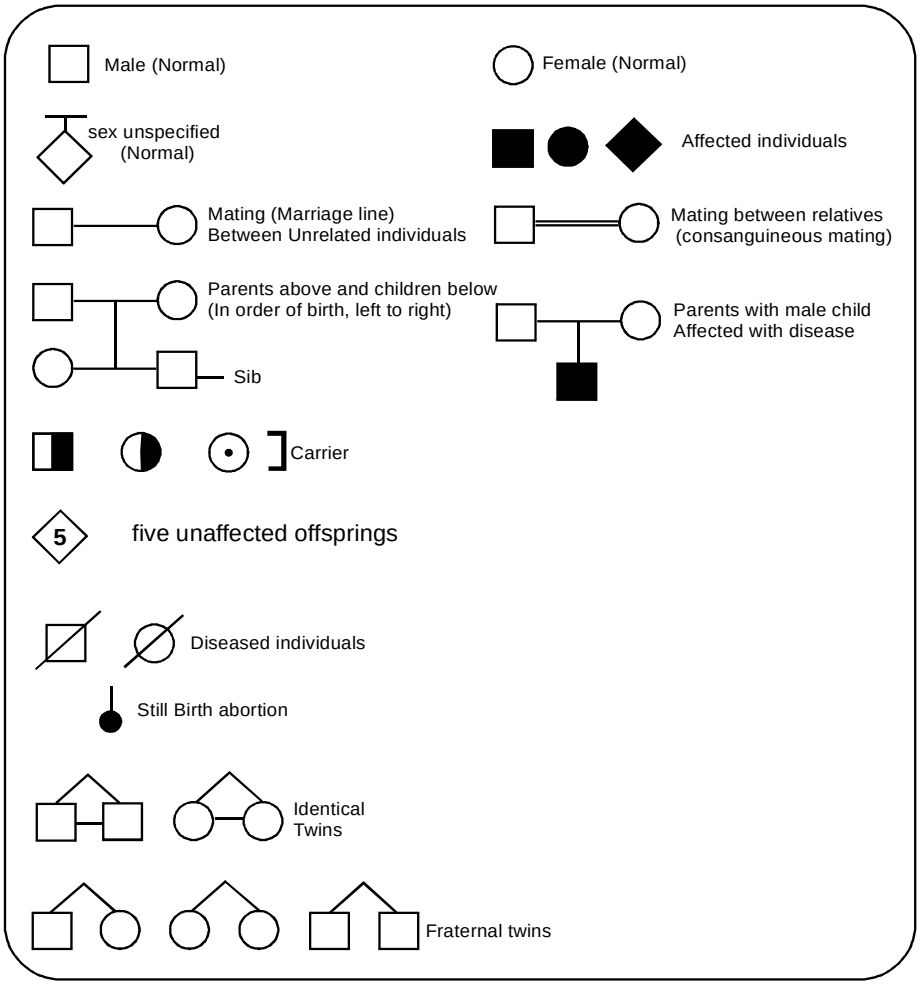
FOR AIIMS & AIPMT :

1. Pedigree Analysis :

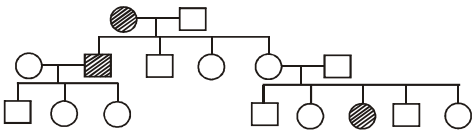
- A record of inheritance of certain genetic traits for two or more generations presented in the form a diagram or family tree is called pedigree. **Eg : Human, domesticated animals**
- Square represents the male  . A circle represents the female  .
- Solid symbol shows the trait under study and a cross or shade of any type  or  , or .

Words can also be used in place of symbols.

- Parents are shown by horizontal line while their offsprings are connected to it by a vertical line.
- Pedigree analysis is study of pedigree for the transmission of particular trait and finding the possibility of absence or presence of that trait in homozygous or heterozygous state in a particular individual.
- It is useful for the genetic counsellors to advice intending couples about the possibility of having children with genetic defects like haemophilia, colourblindness, alkaptonuria, phenylketonuria, thalassemia, sickle cell anaemia, polydactyly & syndactyly.
- Pedigree analysis employs two tools.
  - **(i)** Realised ratio of probability and chances of difference in realised ratio due to smallness of the progeny
  - **(ii)** Elimination of alternatives.



e.g.



The above pedigree chart shows the inheritance of

(1) Colour blindness (2) Huntington corea (3) Alkeptonuria (4) Polydactyly

Ans. (3)

2. Study of Twins: Birth of two babies simultaneously by woman. Twins are of following types.

(i) Dizygotic twins or fraternal twins: If twins develop from two separate fertilized eggs, they are called dizygotic or fraternal twins.

(ii) Monzygote twins: Twins develop from the same fertilized egg (zygote) due to splitting of zygote in to two blastomeres. The former also called identical twins because they are genetically similar except occasional mutation.

(iii) Siamese twins: Sometime breaking of young embryo is incomplete so that monzygotic twins are joined in various regions.

3. Population Genetics:

The study of distribution of traits and frequency of gene distribution in the whole population is called population genetics. It is based on principles of probability and statistical tools.

Hardy-Weinberg law: This principle says that allele frequencies in a population are stable and is constant from generation to generation. The gene pool (total genes and their alleles in a population) remains a constant. This is called genetic equilibrium.

Gene A = p ; Gene a = q ; p + q = 1 or A + a = 1 ; and $(A + a)^2 = A^2 + 2Aa + a^2 = 1$. $1 - \sqrt{q^2} = p$

In a population :

The frequency of dominante allele A $\rightarrow$ p

The frequency of recessive allele a $\rightarrow$ q

Frequency of homozygous dominant individuals AA $\rightarrow p^2$

Frequency of homozygous recessive individuals aa $\rightarrow q^2$

The frequency of heterozygous individuals Aa $\rightarrow$ 2pq

Question : (1) Presence of recessive trait is 16% . The frequency of domimant allele in population will be

Solution

q = $\sqrt{.16}$ = 0.4. p = 1.0 – 0.4 = 0.6

Question: (2) Frequency of an autosomal lethal gene is 0.4. The frequency of carrier in a popoulation of 200 individual will be

Solution

Given q = 0.4 p = 1 – 0.4 = 0.6

Frequency of carrier 2 pq = 2 x 0.6 × 0.4 = 0.48 or 48%

hence 96 out of 200.

Five factors are known to affect Hardy-Weinberg equilibrium. These are gene migration or gene flow, genetic drift, mutation, genetic recombination and natural selection.

Human Genetic Disorders:

(A) Chromosmal Disorders:

(i) Sex Chromosomal Disorders: These are as follows.

(1) Klinefelter's Syndrome:

❖ It is found in man. Its patient contains 47 chromosomes and Its genotype is **44 + XXY**. These persons are sterile males having undeveloped testes. Sparse body hair, mental retardation, and some female characters like **development of breasts (gynaecomastia).** It is due to the presence of additional X-chromosome. It has one barr body. One in every 500 male births is victim of this syndrome.

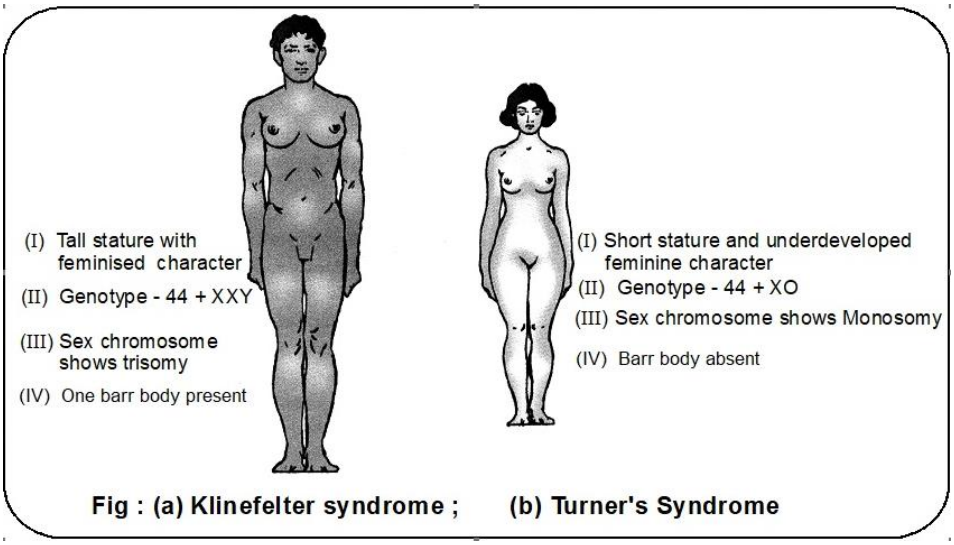


Fig : (a) Klinefelter syndrome ; (b) Turner's Syndrome

(2) Turner's Syndrome:

❖ It is found in woman. The patient has 45 chromosomes instead of 46 and its genotype is **44 + X 0**. These are sterile females having rudimentary ovaries, small uterus undeveloped breasts, short stature, abnormal intelligence, webbed neck. Menstrual cycle is abnormal or absent. One in every 3000 children is a victim of this syndrome.

Resonate the Concept

- * XO- Chromosomal abnormality in human beings causes turner's syndrome. While in some insects like grasshopper, XO type of sex chromosome determines male sex.

(3) Supermales (Jacob's syndrome or criminal syndrome):

❖ The genotype of supermale is **44 + XYY** & total number of chromosomes are 47 instead of 46. These males are characterised by abnormal height, mental retardation and criminal bent of mind **(Jacob's syndromes)** supermales are more aggressive than normal males due to over secretion of male sex hormones.

(4) Superfemale:

❖ The genotype of superfemale is either **44 + XXX (47 chromosomes) or 44 + XXXX (48 chromosomes) or 44 + XXXXX (49 chromosomes).** They have abnormal sexual development and mental retardation.

(ii) Autosomal Disorders: These are as follows.

(1) Down's Syndrome (Mongolian Idiocy):

- It was discovered by **Langdon Down**. In this Syndrome **21-pair of chromosomes** show **trisomy** (presence of an extra chromosome number 21.
- It is caused by nondisjunction of 21st chromosome pair during anaphase. Thus the total number of chromosomes are 47 instead of 46 & genotype is 45 + XY in male & 45 + XX in female.
- Symptoms of this syndrome are broad fore-head, rounded face, permanently open mouth with protruding tongue, projecting lower lip, Mongolian type eye lid fold,verticalfold epicanthus on either side of nose, stubby fingers,broad palm with characteristic palmer crease, Short neck , little intelligence, undeveloped Gonads and genitalia.

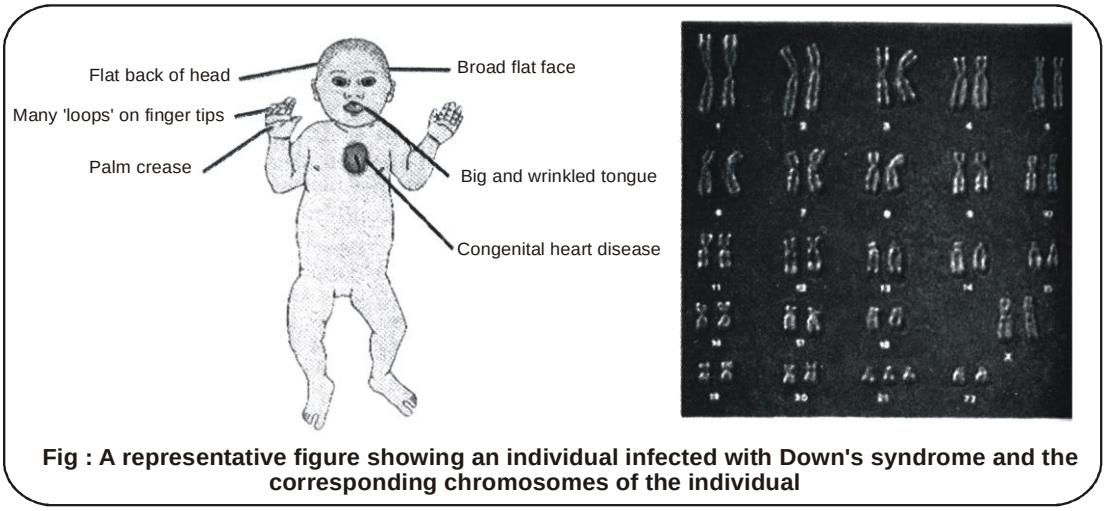


Fig : A representative figure showing an individual infected with Down's syndrome and the corresponding chromosomes of the individual

Resonate the Concept

- **Edward's Syndrome:**
  It is due to **trisomy** in **18th pair of chromosomes**.The affected person keeps the fingers tightly clenched against the palm of the hand. Other symptoms are small jaws, deformed ears, small mouth, nose and fingers, small sternum and pelvis. The patient is mentally retarted and dies within 6 months after birth.
- **Patau's Syndrome :**
  It occurs due to **trisomy** in **13th pair of chromosomes**. It is characterised by small head, abnormalities of the face, eyes and forebrain, cleft lip and palate, deformed ears, small chin and the hands are often clenched as Edward's syndrome. The average life span of the affected person is about 4 months.
- **Cri du chat Syndrome or Cat Cry Syndrome :**
  It is due to **deletion** of half part in the **short arm of the chromosome number 5.** It was reported by **Lejeune (1963)**. The affected newborn cries like mewing of a cat hence it named Cri du chat (Cat Cry). Its other symptoms are moon like face, widely spaced eyes, small head, receding chin, congenital heart disease.
- **Myelogenous Leukemia:**
  It was firstly reported in Philadelphia in 1959 hence it is also called Philadelphia syndrome. It is due to deletion of small part of **long arm of chromosome 22** and its addition to **chromosome 9 (reciprocal translocation).**

(b) Gene Related Disorders: They occur due to alternation or mutation in the single gene. These are as follows.

(i) Gene Mutations in Autosomes: Two types involve in them.

(A) Recessive Traits. **(B) Dominant Traits.**

(A) Recessive Traits: Recessive autosomal genes in homologous condition are responsible for them.

(1) Phenylketonuria (PKU):

- It described by Folling (1934). It is an **autosomal recessive metabolic disorder. Enzyme phenylalanine hydroxylase** is absent due to abnormal autosomal recessive gene on **chromosome 12** as a result phenylalanine (amino acid) is not converted into tyrosine (amino acid) in liver. It is called **hyperphenylalaninemia.** The latter is characterised by accumulation and excretion of phenylalanine, phenylpyruvic acid and related compounds. Symptoms are mental retardation (**IQ less than 20),** decreased pigmentation of hair and skin and eczema.

(2) Alkaptonuria:

- It was **first inborn metabolic disease** explained by **Garrod**. It is an autosomal recessive, metabolic disorder. **Deficiency** of an **alkapton oxidase/homogentisate oxidase enzyme** of liver is responsible for it, as a result **homogentisic acid/Alkapton** accumulates in the tissues and is also excreted in the urine. The latter turns black in the air due to oxidation of homogentisic acid & other symptoms are arthritis, bronz pigmentation.

(3) Albinism:

- Albinos lack dark pigment **melanin** in the **skin, hair** and **iris**. It is an autosomal, recessive genetic disorder. Synthesis of melanin pigment from dihydroxyphenyalanine is absent due to lack of **enzyme tyrosinase**. Only homozygous individual (aa) is affected by this. It is due to recessive allele of **long arm of chromosome 11** but may also be caused by another recessive allele of **P-gene** on long arm of **chromosome 15**.

(4) Cystic Fibrosis:

- The disease is common in Caucasian population. It occurs due to recessive allele of **chromosome number 7**. Symptoms of this disease are the **failure of chloride ion transport mechanism** followed by elevated levels of sodium and chloride in the sweat. Thick mucus accumulates in lungs and respiratory path. It causes blockage and secondary infection. This disorder was formerly called **mucoviscoides**. There is impairment of pancreatic and liver functions in most of the cases of cystic fibrosis. Cardiac failure may occur.

(5) Tay-Sach's Disease (TSD) / Infantile Amourotic Idiocy:

- It is recessive autosomal disorder that occurs due to **deficiency** of **enzyme β-D-N-acetyl hexosaminidase** after birth. Symptoms of this disease involve damaging brain and spinal cord. Mental retardation and paralysis due to **accumulation of lipid GM_2 or Tay-Sach's ganglioside**. The child is dead at the age of 3–4 yrs.

(6) Sickle Cell Anaemia:

- It occurs due to autosomal codominant (formerly considered recessive) allele Hb^s present on **chromosome 11**.

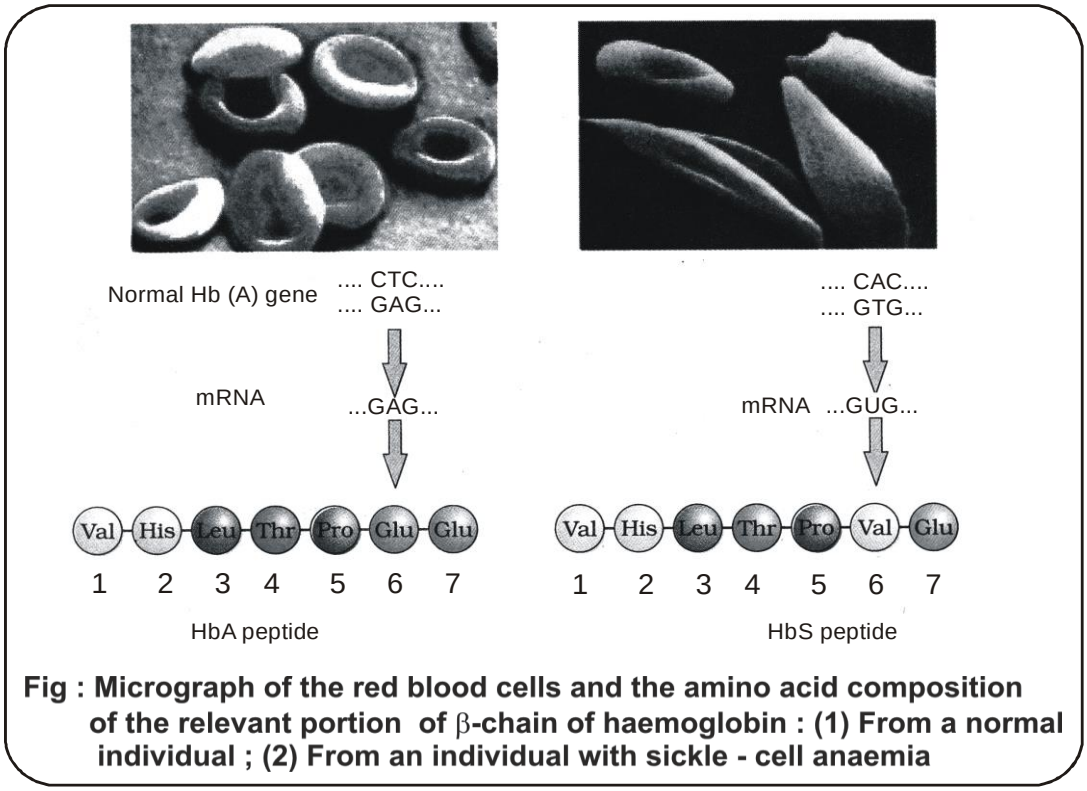


Fig : Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin : (1) From a normal individual ; (2) From an individual with sickle - cell anaemia

- It is caused by the formation of an abnormal **haemoglobin-S**. In which **Glutamic acid**, of **6th position** in **β-chain** is replaced by **Valine** amino acid due to substitution of T by A in the second position of the triplet **codon (CTC)** on chromosome 11.The codon **CTC** usually transcribed into **GAG (coding for Glutamic acid).** but due to substitutions of T by A The new codon **CAC** is transcribed into **GUG** that codes **Valine**. It is the **major effect** of allele.
- **Other secondary effects** involve formation of **sickle-shaped erythrocytes** during oxygen deficiency. The cells cannot pass through narrow capillaries resulting the latter become clogged. Spleen and brain get damaged. The homozygotes having only haemoglobin-S (Hb^SHb^S) usually die before reaching maturity due to erythrocyte distortion but Hb^AHb^S individuals survive.

(B) Dominant Traits:

(1) Huntington's chorea: It is due to **dominant autosomal gene** situated on **chromosome 4,** in which muscle and mental deterioration occurs. Gradual loss of motor control resulting in uncontrollable shaking and dance like movements, slurring of speech, loss of memory and hallucinations after shrinks of brain between 20–30% size. Disease may start at the age of 15– 40 yrs.

(2) Polydactyly: presence of extra fingers and toes.

(3) Achondroplasia: It is a type of dwarfism in which long bones do not grow.

(4) Brachydactyly: abnormal short fingers and toes.

Resonate the Concept

(i) Alzheimer's Disease: It is **neuro-degenerative disease of brain** that occurs due to accumulation of **amyloid protein plaques (amyloid-b peptide protein)** resulting degeneration of neurons takes place in patient. Two defective autosomal alleles,(one on the **chromosome 21** and other on **chromosome 19**) are involved. They produce disease after the age of 40. **Symptoms** involve **changes in personality, memory loss, trembling of hands followed by progressive increase in dementia over next 5–10 yrs.**

(ii) Thalassemia: It is a group of genetic disorders which result from defective synthesis of subunits of haemoglobin. It represent quantitative inheritance. In α-thalassemia, out of the four genes present on 16th chromosomes, absence of four genes (α)-/α- or α α/-- produces microcytic and slight hypochromic erythryocytes without significant anaemia but deficiency of three genes (--/α-) develops microcytic hypochromic erythrocytes with marked haemolytic tendency. Death occurs in foetus in case of deficiency of all the genes. In β-thalassemia the two β-genes present on 11th chromosomes are defective. In β-thalassemia minor (one defective β-gene), the erythrocytes are microcytic with higher RBC count but 15% lower haemoglobin than normal. In β-thalassemia major, both the genes are defective. There is severe anaemia with microcytic hypochromic RBCs.

Cytoplasmic inheritance:

- It was discovered by **Correns (1909) in Plastid inheritance in *Mirabilis jalapa***. The Inheritance of some characters is controlled by cytogene of cytoplasm. It is called cytoplosmic inheritance.
- The genes controlling cytoplasmic inheritance are called **plasma genes or extra-nuclear genes or plasmon** that are found in mitochondria, plastids, plasmids, special particles like kappa particles, sigma particles. male gamete of higher plant has very minute or nil cytoplasm so male gametes only inherit karyogene thus inheritance of cytogene takes place through female.
- It is also called maternal inheritance because zygote receives most of its cytoplasm from the ovum. Thus it is **uniparental**. If there is a reciprocal cross in this condition, then results may be affected.

Resonate the Concept

1. **Fairchild:** It is first successful plant hybrid (fairchild's Mule) that is formed by cross between Dianthus (carnation) and centuria (Sweet William).
2. **Pleiotropic mutation:** Mutation in a gene which has more than one phenotypic expression will affect a variety of characters, such mutation is called pleiotropic mutation **Eg : In sweet pea a single** point mutation cause changes in seed coat colour from Grey to white and flower colour from red to white.
3. **Homeotic Mutation:** A mutation that causes part to form another structure instead of forming its normal structure.
4. **Barr Body:** It was discovered by **Barr & Bertman** in interphase stage of nerve cell of female cats. According to Mary Lyon (1962) one of the two X-chromosomes of a normal female becomes heterochromaticand appears as Barr body. It appears as **drum stick** (these are sex chromatin present in the neutrophil of 3 to 5% cells of females and absent in the neutrophils of males) in leucocytes of females. This inactivation of one of the two X-chromosomes of a normal female is called as dosage compensation or '**Lyon's hypothesis**'. The number of Barr bodies is always less than the number of X- chromosome or X - 1.

Table

S.No	Organism	No. of X – hromosome	No. of Barr body (X – 1)
1	Normal female	XX	2–1= 1 (1 barr body)
2	Normal male	XY	1 –1 = 0 (No barr body)
3	Super female	XXX	3 – 1 = 2 (two barr body)
4	Klinfelter syndrome	XXY	2 –1 = 1 (1 barr body)
5	Turner syndrome	XO	1 –1 = 0 (No barr body)
6	Super male	XYY	1 –1 = 0 (No barr body)

5. **Deficiency of G-6PD:** The enzyme glucose 6- phosphate dehydrogenase is present in RBC for minor glycolysis. The absence of this enzyme is a sex linked recessive trait. The persons deficient in this enzyme cannot have plasmodium in their RBC and hence cannot suffer from malaria. But if such person are given antimalarial drugs, the RBCs, being fragile, can repture and cause severe anaemia.

6. **IQ (Intelligent Quotient):** It is the measurement of intelligence. It is governed by 11 sets of genes. IQ is calculated as.

$$\textbf{IQ} = \frac{\textbf{Mental age}}{\textbf{Actual age}} \times 100$$

Suppose a ten year child has mental age 13, his IQ will be $\frac{13}{10} \times 100 = 130$.

7. **Eugenics:** Improvement of human race by the application of genetics is called Eugenics. **Francis Galton** is called the '**Father of eugenics**'.
8. **Euthenics:** Improvement of the human race by improving the environmental conditions is called Euthenics.
9. **Euphenics:** The symptomatic treatment of genetic diseases of man is called euphenics.
10. **Taste Blindness of PTC:** It was discovered by **Fox (1932)**. It is a dominant genetic trait, PTC (Phenylthiocarbamide) has sour taste. About 30% people lack the ability to taste PTC. Genotype TT and Tt are taster of PTC. tt are nontasters (taste blind persons).