

PRINCIPLE OF INHERITANCE AND VARIATION

- Inheritance: process by which characters are passed on from parents to progeny.
- Variation : degree by which progeny differ from their parents and from one another.
- Genetics : deals with the inheritance, and variation of characters between parents to offspring.

MENDEL’ S LAWS OF INHERITANCE:

- Gregor Johan Mendel – Father of Genetics.
- Performed Plant hybridization experiments on garden peas from 1856 – 1863
- Proposed 3 laws of inheritance based on his experimental findings.

1. Law of Dominance:

- ❖ Characters are controlled by discrete units called factors.
- ❖ Every individual has a pair of factors for each character.
- ❖ When a dissimilar pair of factors is present together in a hybrid, one member of the pair dominates the other (recessive). The allele which dominates the other is called dominant allele while the other which do not express and remain hidden is called recessive allele.

2. Law of Segregation/ Law of purity of gametes:

- ❖ When a pair of contrasting factors or alleles are brought together in a hybrid, the allelic pair remain together without blending and during gamete formation the two separate from each other such that only one allele enters each gamete.

3. Law of independent Assortment:

- ❖ When two characters (dihybrid) are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.

Genetic Terms

- ❖ **Factors:** Hereditary unit that was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations is termed as ‘factors’ (now called **genes**).
- ❖ **Character** – Feature of an individual, like Height, colour, shape etc.
- ❖ **Trait** - a specific form of a character, like Tall and dwarf of height character.
- ❖ **Alleles** = Contrasting forms a gene defining different traits of a character. They occupy same loci on homologous chromosomes.
- ❖ **Phenotype** – morphological/physical appearance
- ❖ **Genotype** – total set of alleles present in an organism
- ❖ **Homozygous** – similar alleles of a gene in an individual
- ❖ **Heterozygous** – dissimilar alleles of a gene in an individual

Understanding gene, alleles, dominant and recessive

- **Gene:** A segment of DNA that contains the information for producing an enzyme.
- **Allele:** There are two copies of a gene, present at same loci on homologous chromosomes. These two copies of a gene are the two allelic forms of the gene.
- **Dominant allele:** The normal gene producing the normal enzyme that is needed for the transformation of a substrate S.
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- Recessive allele: The modified gene could be responsible for production of –
 - (i) the normal/less efficient enzyme, or
 - (ii) a non-functional enzyme, or
 - (iii) no enzyme at all

MENDEL'S EXPERIMENT (1856-1863)

- Artificial/cross pollination using several true-breeding 'pea' lines (one having undergone continuous self-pollination for several generations).
- Characters selected - 7 contrasting traits.

Character	Dominant trait	Recessive trait
1. Stem height	Tall	Dwarf
2. Flower colour	Violet	White
3. Flower position	Axial	Terminal
4. Pod shape	Inflated	Constricted
5. Pod colour	Green	Yellow
6. Seed colour	Yellow	Green
7. Seed shape	Round	Wrinkled

MONOHYBRID CROSS:

- Cross between two varieties having a contrasting character.
- Experiment to observe the inheritance of a pair of alleles controlling different forms of a character.

Experiment:

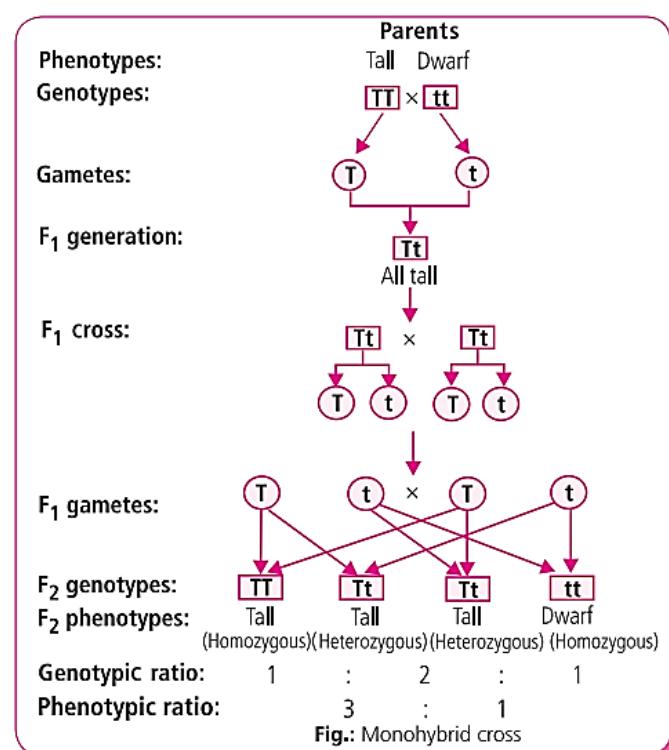
- Artificial hybridisation of pure lines varieties. It involves emasculation and bagging.
e.g. Tall and dwarf plants.
- Hybrid seeds sown to raise First hybrid generation (filial 1 progeny or the F₁).
- Self pollination of F₁ hybrids give F₂ generation.

Result of monohybrid cross:

- ❖ F₁ progenies always have trait of only one of the parents.
- ❖ The trait that appears in F₁ is termed as **dominant trait** while trait that does not appear in F₁ is termed as **recessive trait**.
- ❖ In F₂, the recessive trait is expressed in 1/4th of the offspring and dominant trait in 3/4th of the progenies.
- ❖ i.e. F₂ Phenotypic ratio = 3:1
- ❖ All offspring identical to their parents.
- ❖ No blending or intermediate character at either F₁ or F₂ stage.

Modern Genetics and explanation:

- ❖ Every individual has a pair of alleles for a character.



- ⊕ Dominant alleles = denoted by capital letter
- ⊕ Recessive allele = denoted by small case of dominant letter.
- ❖ Law of Dominance & Law of Segregation – laws to explain the observations of monohybrid cross:
 1. Expression of only one of the parental traits in F1 in a monohybrid cross.
 2. Expression of both parental traits in F2.
 3. The 3:1 ratio of parental phenotypes in F2.

BACK CROSS

- Crossing of a hybrid(a first-generation hybrid) with one of its parents
- The quickest way to generate homozygous offspring.

TEST CROSS

- Cross between an unknown (F1 hybrid) with Recessive parent i.e. Tt x tt
- To test the genotype of unknown parent E.g. Tall can have two genotypes = TT or Tt
- Test cross identifies the genotype of unknown plants.

INHERITANCE OF TWO GENES or DIHYBRID CROSS:

- Crosses between plant varieties differing in two traits.
- The cross is done to observe the inheritance two characters.

Result

- New recombinant phenotypes appear in F2.
- Phenotypic ratio = 9:3:3:1
- Genotypic ratio = 1:2:1:2:4:2:1:2:1
- Parental/recombinant = $\frac{10}{6}$
- Parental to recombinant genotypic ratio = 2:14 = 1:7

Punnet square:

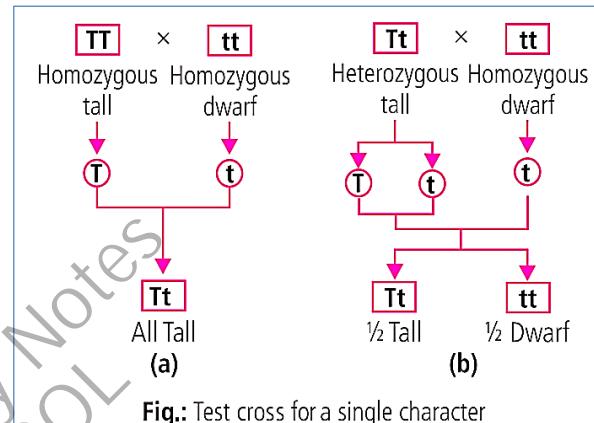
- Graphical representation to calculate the probability of possible genotypes in a genetic cross.
- Developed by Reginald C. Punnett – British Geneticist.

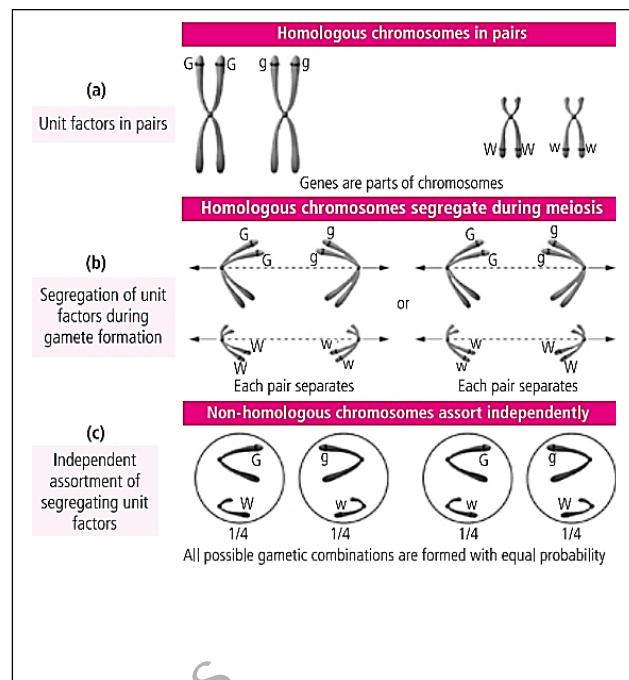
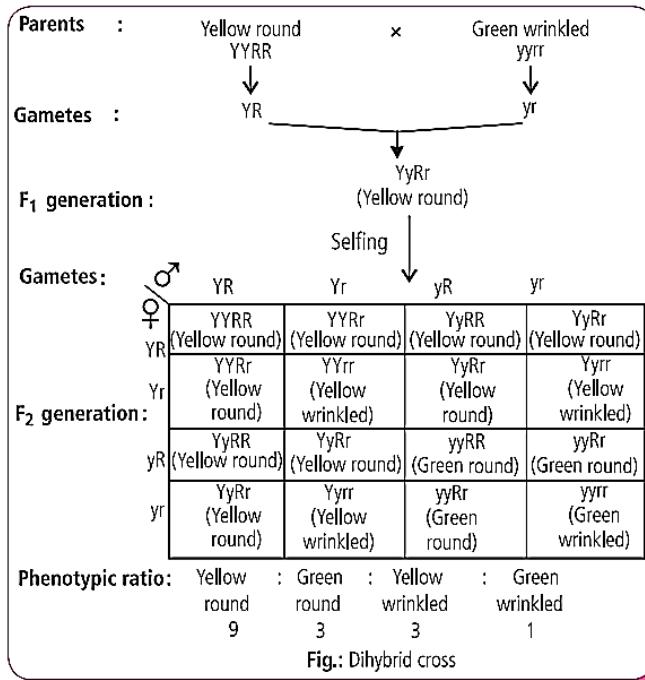
Law of independent Assortment:

- When two characters (dihybrid) are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.

Explanation:

- RrYy (F1 hybrid) produces four types of gametes - RY, Ry, rY, ry in equal frequency(25%).
- This is because segregation of R and r is *independent* from the segregation of Y and y i.e. it is a random process (2nd law).
 - Therefore, R bearing gametes has 50/50 chance of having Y or y.
 - Similarly, r bearing gametes has 50/50 chance of having Y and y.





- ❖ Mendel's F₂ 9:3:3:1 dihybrid ratio is an ideal ratio based on probability events involving segregation, independent assortment, and random fertilisation.
- ❖ It is applicable to only those factors or genes which are present on different chromosomes.

Reasons for Mendel's Success:

- Application of Statistical analysis and mathematical logic to solve problems in biology.
- A large sampling size, which gave greater credibility to the data.
- Confirmation of his inferences from experiments on successive generations.
- Use of garden pea plant that manifested characters as two opposing traits.

Why Mendel's theory was remained unrecognized?

- i) Firstly – communication was not easy in those days and his work could not be widely publicized.
- ii) Secondly – his concept of factors as stable and discrete units that controlled the expression of traits which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.
- iii) Thirdly – Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.
- iv) Finally – he could not provide any physical proof for the existence of factors.

Rediscovery of Mendel's result:

- 1990 three scientists
 - i) Hugo de Vries of Holland
 - ii) Carl Correns of Germany
 - iii) Erich von Tschermak of Austria
 - independently rediscovered Mendel's result on the inheritance of character.

Deviations from Mendelian Inheritance

A. Incomplete dominance:

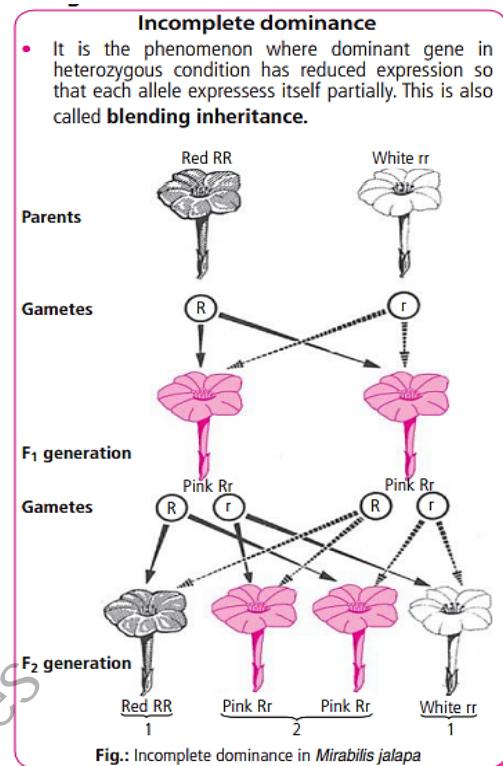
- ⊕ It is the genetic phenomenon in which a dominant allele does not completely mask the effects of a recessive allele.
- ⊕ This results in a third phenotype in F₁ hybrid which is intermediate of the two.
- ⊕ E.g. :
 - i) Inheritance of flower colour in the dog flower (snapdragon or *Antirrhinum sp.*)
 - Both phenotypic and Genotypic ratio is 1:2:1 i.e. is equal to one another.
 - ii) Starch grain size in pea
 - a. BB – Large grain
 - b. Bb – Intermediate
 - c. bb - small

B. Co – dominance:

- ⊕ It is genetic phenomenon in which two alleles express themselves independently when present together in an organism.
- ⊕ Joint expression of both alleles in F₁ hybrid.
- ⊕ F₁ hybrid resembles both parents side by side.

Example 1. ABO blood grouping in human

- Controlled by the gene *I* with three alleles I^A, I^B and i where I^A and I^B are completely dominant over i.
- Gene - I produce sugar polymers (antigen) that protrude from plasma membrane of the RBC. This decides blood group.
- Each person possesses any two of the three I gene alleles.
 - I^A and I^B produce a slightly different form of sugar
 - i doesn't produce any sugar.
 - I^AI^B = both sugar present = co-dominance.
- Example 2. Roan character in cattle



INCOMPLETE DOMINANCE	CO-DOMINANCE
• The condition when neither of the alleles is dominant, rather combine and display a new trait by mixing of the two alleles	• The condition when both the alleles of a gene are dominant, and the traits are equally expressed
• The hybrid will always give rise to new phenotype.	• No formation of the new phenotype.
• Though both the allele blend their effect, one of the two is more noticeable.	• Here both the alleles equally blend and show their equal effects.
• Snapdragon, <i>Mirabilis jalapa</i>	• Roan character in cattle, A and B blood group in human

C. Multiple Alleles:

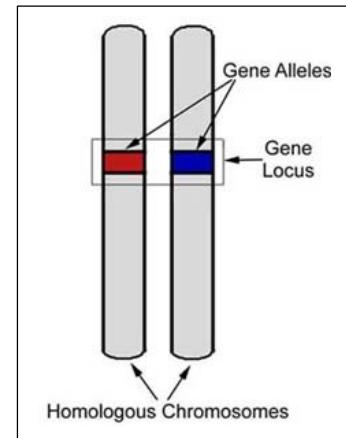
- Presence of more than two alleles in a loci, governing same character.

E.g.: ABO blood group.

- Control by the gene I with three alleles I^A , I^B and i .
- A diploid individual can have at the maximum of two alleles at the same gene locus (on homologous chromosomes).
- Hence multiple allele is observed in population study only, not in an individual.

- ❖ The alleles controlling the ABO blood group thus shows
- Complete dominance
 - Incomplete dominance
 - Multiple allelism

Phenotype (Blood group)	Genotypes	Antibodies present in blood serum	Antigens on RBC
A	I^A/I^A or I^A/i	B	A
B	I^B/I^B or I^B/i	A	B
AB	I^A/I^B	None	A and B
O	ii	A and B	None



D. Pleiotropy:

- It is the phenomenon in which a single gene can exhibit multiple phenotypic expression.
- Such gene is called **pleiotropic gene**.
- In most cases the gene affects metabolic pathways which lead to different phenotypes.

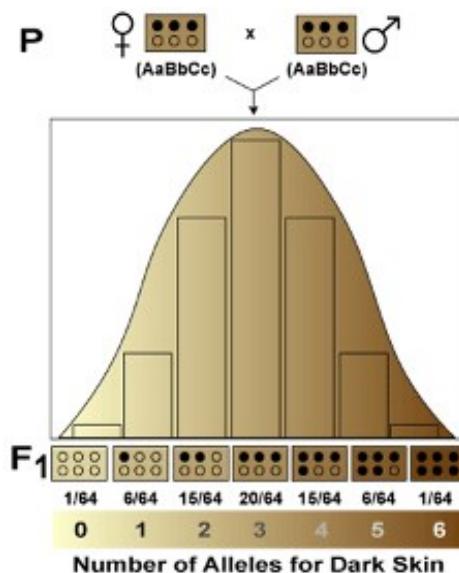
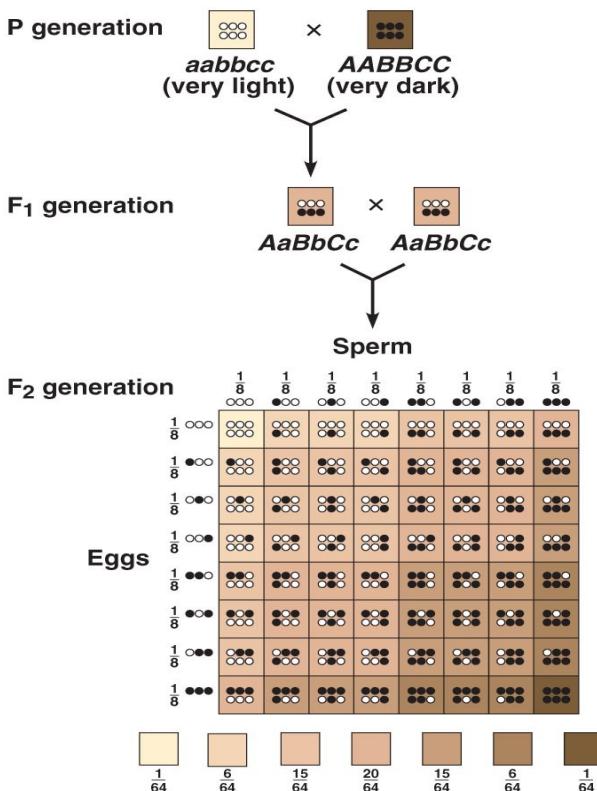
- E.g.
- Phenylketonuria a disease in human is an example of pleiotropy.
 - Arise due to mutation in the gene encoding phenyl alanine hydroxylase.
 - Phenotypic expression characterized by-
 - Reduction in skin and hair pigmentation.
 - Mental retardation due to accumulation of metabolites.
 - Gene controlling starch grain size also control shape of seed in pea

E. Polygenic/Quantitative Inheritance:

- Inheritance of traits controlled by three or more genes are called **Polygenic inheritance**
- Origin of polygenic trait.
 - Duplication of chromosome part
 - Increase in chromosome number
 - Mutations producing genes having similar effect.

E.g.

- skin colour = under control of Three genes A, B, C.
 - Dominant forms A, B, C = dark skin colour
 - Recessive forms a, b, c = light skin colour.
- Phenotypic effect of each allele is additive i.e. more dominant allele more intensity of the trait.
- Hence show continuous variation.
 - $AABBCC$ = dark.
 - $aabbcc$ = light.
 - $AaBbCc$ = Mulato



Important Formulae

- ✚ Type of gametes / phenotypic category = 2^n
- ✚ Type of genotype = 3^n , (n = no of heterozygous pair)
- ✚ No of progeny = no of male gametes x no. of female gamete.

Deviations from Mendelism

	Mendel's Observations	Deviations
1.	A single gene for a single phenotype	Pleiotropy: Single gene exhibits multiple phenotypic expressions
2.	Only of the parental characters appear in a monohybrid cross in F ₁ generation.	Incomplete dominance: In a monohybrid cross phenotype in the F ₁ generation do not resemble either of the two parents and was in between the two
3.	Only of the parental characters appear in a monohybrid cross in F ₁ generation.	Co-dominance: In a monohybrid cross, phenotype in the F ₁ generation resemble both the parents
4.	All the characters have distinct alternate forms since one gene is responsible for one character.	Polygenic Inheritance: Occurrence of a range of forms of a character in a population. Such characters are controlled by two or more genes.

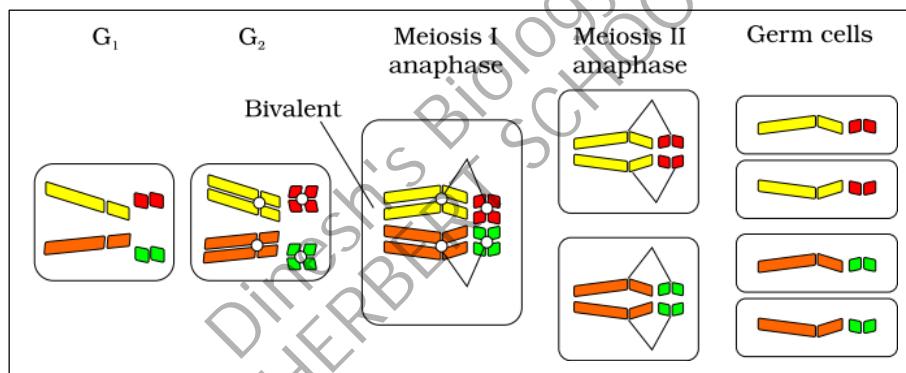
CHROMOSOMAL THEORY OF INHERITANCE:

Chromosomal theory of inheritance:

- Proposed by Walter Sutton and Theodore Boveri in 1902.
- They observe parallelism between chromosomes and Mendelian factors.
- Separation of chromosomes during meiosis could serve as the cytological basis of Mendel's law.

Chromosomal theory of inheritance states that

- i) Chromosomes are the carriers of genetic material.
- ii) Genes which control characters are arranged linearly along the length of chromosomes.
- iii) Chromosome like genes are present in pairs in diploid cells.
- iv) Like alleles, homologous chromosomes paired and separate during gamete formation (meiosis).
- v) The pairing and separation of chromosomes would lead to the segregation of character pair in offsprings.
- vi) Fertilization restores the chromosome number to diploid condition.
- vii) The segregation and random assortment of chromosomes are the basis for all genetic inheritance.



- ❖ The genes controlling the seven pea characters studied by Mendel are now known to be located on four chromosomes (1, 4, 5, 7).

Chromosome no.	Character present.
chromosome no 1	1.seed coat
chromosome no 4	2.seed cotyledon colour. 3.plant hight 4.position of flower.
chromosome no 5	5.pod shape.
chromosome no 7	6.pod colour. 7.seed shape.

Linkage and Recombination

- Thomas Hunt Morgan, Alfred H. Sturtevant, Calvin Bridges, and others established beyond a reasonable doubt that Sutton and Boveri's hypothesis was correct.
- Morgan worked with tiny fruit flies, *Drosophila melanogaster*.

Why Drosophila?

- Suitable for genetic studies.
- Grown on simple synthetic medium in the laboratory.
- They complete their life cycle in about two weeks.

- A single mating could produce a large number of progeny flies.
- Clear differentiation of male (small) and female (larger) flies.
- Have many types of hereditary variations that can be seen with low power microscopes.

Linkage:

- Tendency of genes present on a chromosome to inherit together in subsequent generations.
- T.H. Morgan and his colleagues, C.B. Bridges and Sturtevant proposed ‘*The chromosome theory of linkage*’ .
- It states that :
 - Linked genes occur in the same chromosome.
 - They lie in a linear sequence in the chromosome.
 - There is a tendency to maintain the parental combination of genes except for occasional crossovers.
 - Strength of the linkage between two genes is inversely proportional to the distance between the two, i.e., two linked genes show higher frequency of crossing over if the distance between them is higher and low frequency if the distance is small.

i.e. Linkage $\propto \frac{1}{\text{Distance between genes}}$
- Genes that are present on the same chromosome make one **linkage group**
- Genes located closely on same chromosome will deviate from 9:3:3:1 ratio or will deviate from dihybrid test cross ration of 1:1:1:1.

Linkage types:

Complete linkage

Complete linkage is a linkage or grouping of genes on a chromosome which is not altered and is inherited as such from generation to generation without any cross-over. In such cases, linked genes do not separate to form new or non-parental combinations.

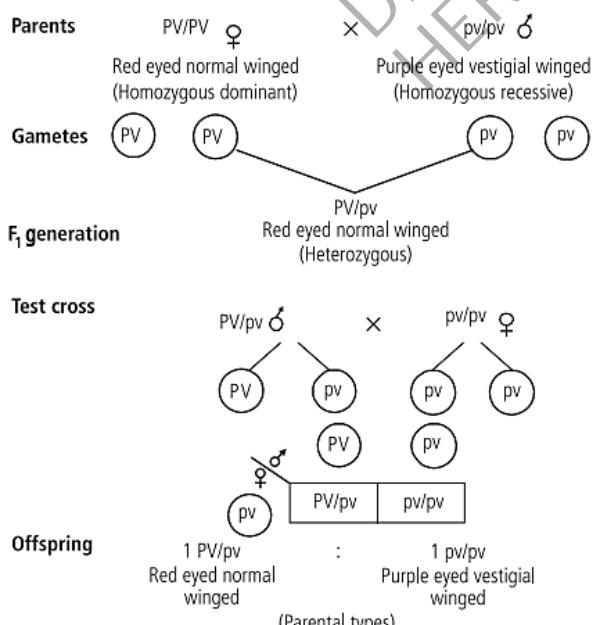


Fig.: Cross showing complete linkage in *Drosophila*

Incomplete linkage

Incomplete linkage is the phenomenon of an occasional crossing over between two homologous chromosomes so that one or more alleles present in a linkage group are replaced by other alleles.

It produces both parental and recombinant individuals. The percentage of each parental type is more than 25% while that of each recombinant type is less than 25%, i.e., parental types are more than 50% of population while recombinant types are less than 50%.

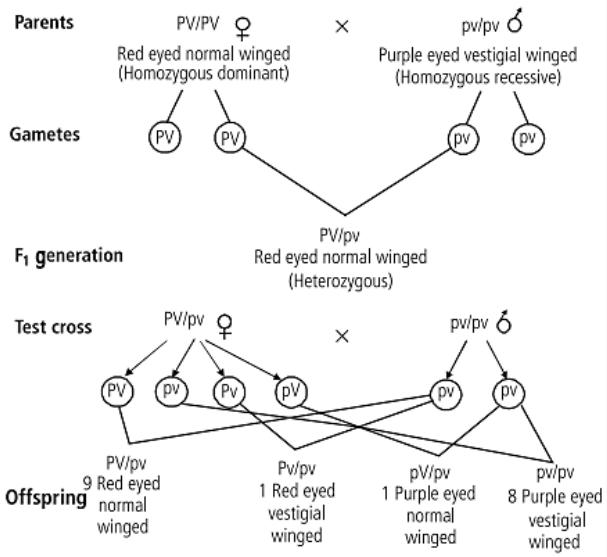


Fig.: Cross showing incomplete linkage in *Drosophila*

Complete linkage	Incomplete linkage
- Completely linked genes always inherit together in subsequent generations	- Link genes having the tendency for occasional segregation in progenies
- They are present very close to one another	- Are distantly located at same chromosome
- No crossing over takes place between them	- Crossing over generate new recombinants
- <i>Progenies have only parental characters</i>	- Progenies show more parental combination along with few recombinants
- E.g. completely linked genes is rare. Genes located on Y chromosome	- E.g. it is more common.

Recombination and crossing over:

- **Recombination** – Generation of non-parental gene combinations in progenies.
- Recombination is due to crossing over
- **Crossing over** – exchange of corresponding chromosome segments between non-sister chromatids of homologous chromosome during meiosis.
- **Steps of crossing over**
 - i) Synapsis – pairing of homologous chromosome
 - ii) Splitting of bivalent to tetrad
 - iii) Breakage and reunion of non-sister chromatids – chiasma formation

Difference Linkage and Crossing over

Linkage	Crossing over
- Tendency of genes in a chromosome to inherit together	- Exchange of chromosome segments to break linkages and establish new linkages
- Produces parental phenotypes	- Frequency increase as if the genes are widely present on chromosome
- Strength of linkage increase if they are closely placed in a chromosome	- Produces new gene combinations

Linkage Map

- Linear graphical representation of the sequence and relative distances of various genes present in a chromosome.
- Alfred Sturtevant developed first “chromosome map” of *Drosophila*.
- Construction of linkage map is based on
 - i) Genes present on the same chromosome are arranged in linear order
 - ii) Recombination between genes is directly proportional to the distance between the two

- Incompletely linked genes showing recombination are used for construction of linkage map
- Frequency of recombination = $\frac{\text{No of recombinant progeny}}{\text{Total number of offsprings}} \times 100$
- Map unit = centi morgan (cM)
- 1 cM = 1% recombination.

Linkage between genes can be observed in dihybrid test cross results.

- ❖ The phenotypic and genotypic ratio will deviate from normal 1:1:1:1 ratio.
- ❖ More parental combination = linkage

Example 1. Cross between AaBb x aabb, F₂ progenies with corresponding genotypes are aabb = 524, Aabb = 206, aaBb = 230, AaBb = 680. What is the distance between A and B of a and b if both located on same chromosome?

$$\text{Ans. R.F.} = \frac{(206+230)}{(524+680)} \times 100 \\ = 36.2$$

Distance = 36.2 cM

Example 2. If recombination frequencies between four genes are

- i) A & B = 3
- ii) B & C = 10
- iii) C & A = 7

Find out the order of the three genes on linkage map

SEX DETERMINATION:

- Biological system that determines the development of sexual characters in organisms.
- Henking (1891) traced specific nuclear structure during spermatogenesis of some insects.
- 50 % of the sperm received these specific structures, whereas 50% sperm did not receive it.
- Henking gave a name to this structure as the X-body.
- X-body of Henking was later on named as X-chromosome.
- Chromosome types
 - Aotosome = body characters
 - Allosome = sex determining genes (X,Y in human)

A. Chromosomal Sex determination

- a. **XX-XY type:** e.g. *Drosophila*, Mammals
 - male and female has same number of chromosomes.
 - Female = AA + XX, Gamete = A+X ; homogamety
 - Male = AA + XY, Gamete = A+X, A+Y ; Heterogamety
- b. **XX-XO type:** E.g. Grass hopper, Round worm, Cockroach
 - Female has one chromosome more than male.
 - Female = AA+XX; homogamety
 - Male = XO; Gamete = A+X, A+O ; 50% do not have sex chromosome, heterogamety
 - Fertilization
 - (A+X) & (A+X) = AA+XX = female
 - (A+X) & (A+O) = AA+XO = male

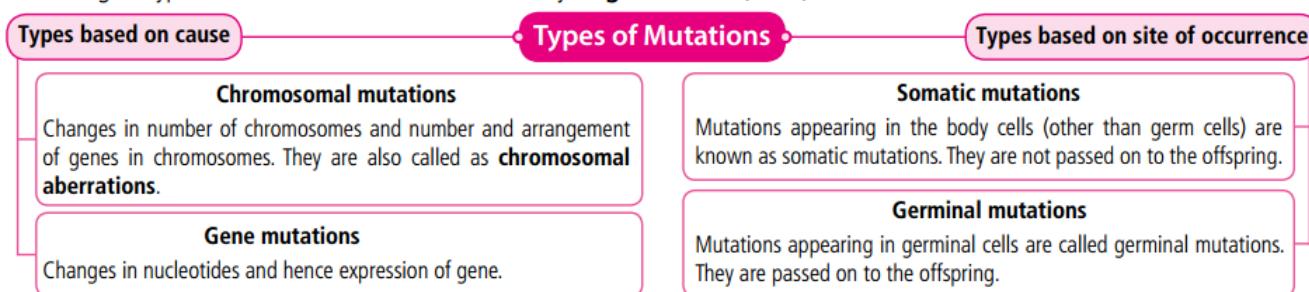
- c. ZW-ZZ type: Birds, fishes, some amphibians
 - male and female has same number of chromosomes.
 - Female = AA + ZW, Gamete = A+Z, A+W ; heterogamety
 - Male = AA + ZZ, Gamete = A+Z; homogamety.
- d. ZO-ZZ type: E.g moths butterfly
 - male has one chromosomes more than female.
 - Female = AA + ZO, Gamete = A+Z, A+O ; heterogamety
 - Male = AA + ZZ, Gamete = A+Z; homogamety.
- e. Sex determination in Honey bee:
 - It is based on the number of sets of chromosomes an individual.
 - Diploid = female (queen and worker)
 - Haploid = Male (drone)
 - Male develops from unfertilized egg through parthenogenesis. Produce sperms by mitosis.
 - Fertilized egg – diploid larvae – feeds on royal jelly – queen (fertile female)
 - Bee bread – worker (sterile female)
 - This is called **haplodiploid sex determination system**.
 - Drones do not have father and thus cannot have sons, but have daughters and grandsons.

B. Environmental sex determination

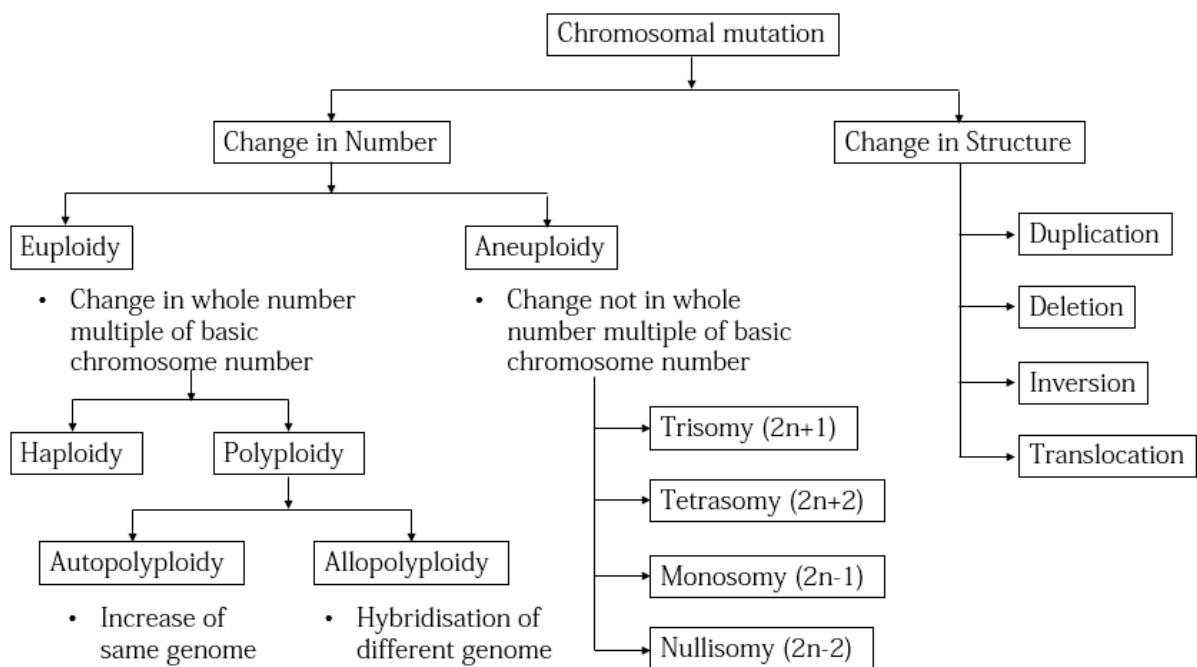
- a. Crocodiles – low temperature – femaleness, high = maleness
- b. Marine fish Medusa – cold water – male, warm water = female
- c. Turtles – below 28C = male, above 33 = female, between = 50-50
- d. *Bonellia* – larva settles alone = female, settles in contact with female = male

MUTATION:

- Mutation is alteration of DNA sequences.
- It results in changes in the genotype and phenotype of an organism.
- In addition to recombination, mutation is another phenomenon that leads to variation in DNA.
- Mutations are new sudden, inheritable, discontinuous variations which appear in the organisms due to permanent change in their genotypes. The term "mutation" was coined by **Hugo de Vries** (1901).



Gene mutations	Inversion	Substitution	Frame shift
	The base sequence of a cistron is reversed.	One base is substituted by other.	Reading of the frame of base sequences shifts laterally.
Transition One purine is replaced by other purine and one pyrimidine is replaced by other pyrimidine.	Transversion Purine is replaced by pyrimidine.	Insertion One or more nucleotides are added in the segment of DNA representing cistron.	Deletion One or more nucleotides are lost from a segment of DNA representing a cistron.



Important points

- Loss (deletion) or gain (insertion/duplication) of a chromosome segment results in change in DNA sequence.
- Since genes are located on the chromosome, alteration in chromosomes results in abnormalities.
- Chromosomal aberrations are commonly observed in cancerous cells.

GENETIC DISORDERS:

MENDELIAN DISORDER:

- Genetic disorders grouped into two categories –
 - ✚ Mendelian disorder
 - ✚ Chromosomal disorder
- Mendelian disorders are due to mutation in the single gene.
- They obey the principles of Mendelian inheritance.
- Can be expressed in pedigree analysis.
- E.g. Haemophilia, Colour blindness, Cystic fibrosis, Sickle cell anaemia, Phenylketonuria, Thalassemia etc.

Types

1. Sex linked disorder – defective gene on X or Y chromosome.
 - E.g. Haemophilia, Colour blindness
2. Autosomal disorder – defective gene on autosomes
 - E.g. Cystic fibrosis, Sickle cell anaemia, Phenylketonuria, Thalassemia

A. Haemophilia (Bleeders disease):

- protein involved in the clotting of blood is affected.
- a simple cut will result in non-stop bleeding in affected individual.

- Two types :
 - Haemophilia A: Factor VIII (Antihaemophilic globulin) (AHG)
 - Haemophilia B : Factor IX (Plasma thromboplastin) Christmas disease.
- Sex linked recessive disease – gene present on X and is recessive.
- Possible genotypes
 - X^hX = Carrier female
 - X^hX^h = Affected female
 - X^hY = Affected male
- Female becoming haemophilic is extremely rare because mother of such a female should be at least carrier and the father should be haemophilic.
- Foetus generally aborted during development.

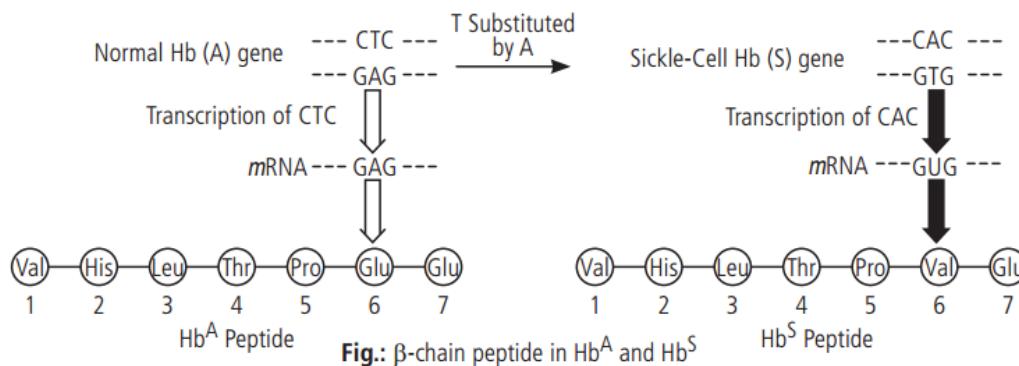
B. Colour blindness

- It is a sex-linked recessive disorder.
- defect in either red or green cone of eye results in failure to discriminate between red and green colour.
- more prevalent in males than females.
- because in males one defective gene is hemizygous and Y chromosome doesn't have corresponding gene.
- In females the disorder is present only in homozygous recessive condition.
- Females with heterozygous condition are unaffected but act as carriers.

1. A male human is heterozygous for autosomal genes A and B and is also hemizygous for haemophilic gene h. What proportion of his sperms will be abh? [2004]
 - 1/8
 - 1/32
 - 1/16
 - 1/4
2. A colour blind man marries a woman with normal sight who has no history of colour blindness in her family. What is the probability of their grandson being colour blind?
 - 0.25
 - 0.5
 - 1
 - Nil[2015]

C. Sickle cell anaemia:

- The defect is caused due to substitution of Glutamic acid (Glu) by Valine (Val) at the 6th position of the β -globin chain of the haemoglobin molecule.
- Substitution is due to transversion of A with T in sixth codon i.e. GAG to GTG.



- The defective haemoglobin
 - polymerization under low oxygen tension
 - leads to change in the shape of the RBC to elongated sickle like structure.

- This is an autosomal recessive trait with defective allele Hb^S.
- Possible genotypes
 - Hb^S Hb^S = affected individual (both male and female)
 - Hb^A Hb^S = carrier and unaffected (both male and female)
- A qualitative problem of synthesising malfunctioning β globin.

D. Phenylketonuria:

- Autosomal recessive trait.
- Inborn error of metabolism.
- The affected individual lack an enzyme called phenylalanine hydroxylase that converts the a.a. phenylalanine to tyrosine.
- phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives.
- Accumulation of these affects brain functioning leading to mental retardation.
- These derivatives excreted through kidney.

E. Thalassemia:

- Autosomal recessive trait.
- Developed due to reduce rate of synthesis of one of the globin chains (α and β chains) of haemoglobin.
- Abnormal haemoglobin molecules results into anaemia – characteristics of the disease.
- Types:
 - ◆ α thalassemia
 - ✓ production of α chain is affected.
 - ✓ controlled by two closely linked genes HBA1 and HBA2 on chromosome 16.
 - ✓ mutation in one or more of the four genes leads to disorder.
 - ◆ β thalassemia
 - ✓ Production of β chain is affected.
 - ✓ Controlled by a single gene HBB on chromosome 11.
 - ✓ mutation in one or two genes leads to the disorder.
- Thalassemia is a quantitative problem of synthesising too few globin molecules

- ❖ Inborn errors in amino acid metabolism : Phenylketonuria, Albinism
 - ❖ Disorders due to abnormal haemoglobin : Sickle cell Anaemia, Thalassemia

Sex linked inheritance:

Sex linkage

- X and Y chromosome = homeologous, Y do not have most of genes present in X.
- Thus genes in X chromosome exhibit unique pattern of inheritance unlike autosomal genes.
- Morgan discovered first in *Drosophila*.

X-linkage

- Inheritance of genes present on allosomes.
- Defect in such genes leads to disorders.
E.g. Haemophilia, colour blindness,

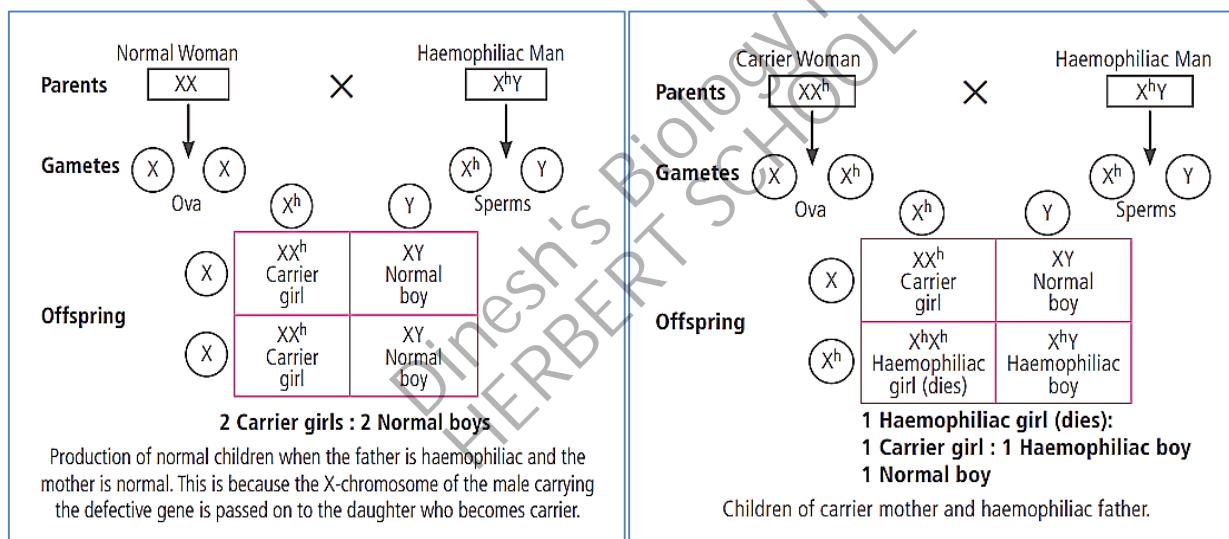
- Sex linked genes are generally recessive.
- Possible genotype combination
 - X^*X = Carrier female
 - X^*X^* = Affected female
 - X^*Y = Affected male

Characteristics of X-linked inheritance

- It shows criss-cross inheritance: father does not pass X-linked trait to his son but the same is passed to grandson through daughter.
- Mother passes the X-linked alleles to both sons and daughters
- Sons always receive the disease from mother not from father.
- X-linked traits are more apparent in males than in females.
- Females generally function as carriers because most of X-linked alleles are recessive and express in females only in homozygous state.

Y-linkage

- Genes of somatic character located on Y chromosome.
- Such genes are called **Holandric genes**.
- e.g. TDF (Testes determining factor)



Chromosomal disorders

- Caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- Source of absence or excess chromosome
 - i) **Nondisjunction:** Failure of segregation of
 - homologous chromosomes or
 - ii) **Failure of cytokinesis:** after telophase stage of cell division results in **polyploidy**.
 - **Trisomy:** additional copy of a chromosome ($2n+1$).
 - **Monosomy:** Lack one of any one pair of chromosomes ($2n-1$)

A. Down syndrome:

- Caused due to presence of an additional copy of the chromosome 21 (trisomy of 21).
- This disorder was first described by Langdon Down (1866).
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- Symptoms

- Short stature with small round head.
- Furrowed tongue
- Partially opened mouth
- Palm is broad with characteristic palm crease.
- Physical, psychomotor and mental development is retarded.

B. Turner's syndrome:

- Caused due to the absence of one of the X- chromosomes i.e. 45 (44 + X0).
 - Such females are sterile as ovaries are rudimentary.
 - Lack of other secondary sexual characters.

Klinefelter's syndrome:

- Caused due to the presence of an additional copy of X-chromosome (44+XXY or trisomy of sex chromosome).
 - Long stature due to long bones.
 - Characteristic web neck.
 - Also develop feminine character (development of breast i.e. Gynaecomastia).
 - Individuals are sterile

PEDIGREE ANALYSIS:

- Analysis of traits in several of generations of a family is called the pedigree analysis.
- In the pedigree analysis the inheritance of a particular trait is represented in the family tree over generations.

Significance

- Helps to determine genotypes by observing phenotypes
- Used by genetic counsellors to advice couples regarding the possibility of children with genetic defect.
- Indicates origin of the trait and consequences of marriage between close relatives.

Symbols used in pedigree chart

