PRINCIPLES OF INHERITANCE AND VARIATIONS

INTRODUCTION

Genetics term was given by **W. Bateson**.

Genetics = Collective study of heredity & Variations.

Heredity = Transmission of genetic characters from parent to offsprings.

Variation = individuals of same species have some differences, these are called variation.

Inheritance :- Process by which character are passed on from parent to progeny.

G.J. Mendel – Father of Genetics.

W. Bateson – Father of Modern Genetics.

Morgan – Father of Experimental genetics

Some importance Definition :-

1. **Factor/Gene :-** Unit of heredity responsible for inheritance of character and appearance of character.
2. **Allele :-** Alternative form of gene. 

3. **Homozygous :-** A zygote is formed by fusion of two gametes having identicle factors is called homozygote and organism developed from this zygote is called homozygous. Ex. TT, RR, tt
4. **Heterozygous** :- A zygote is formed by fusion of two different types of gamete carrying different factors is called heterozygote (Tt, Rr) and individual developed from such zygote is called heterozygous.
5. **Phenotype** :- It is the external and morphological appearance of an organism for a particular character.
6. **Genotype** :- The genetic constitution or genetic make-up of an organism for a particular character.

MENDELISM

Gregor Johann Mendel (1822 - 1884) :- Worked on Pisum Sativum (garden pea 2n = 14)
Working year 1856-1863 (7 years)
Paper : Experiment in plant hybridization

In 1900, Mendel's postulates were rediscovered by three scientists independently. ie.

1. **Carl Correns -** Germany
2. **Hugo deVries -** Holland
3. **Erich von Tschermak Seysenegg -** Austria

Reason for Mendle's Failure :-

(i) Communication was not easy.

(ii) His concept of **genes** (or **factors**, in Mendel's words) was not accepted by his contemporaries.

(iii) Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.

(iv) He could not provide any physical proof for the existence of factors.

Reasons for Mendel's success :

(i) Mendel studied the inheretance of one or two characters at a time.

(ii) Selection of Material –

Selection of garden Pea plant is suitable for studies ;which have the following advantages :

(a) Short life cycle of 2-3 months.

(b) It has many contrasting traits.

(c) Natural self pollination and artificial cross pollination are present in pea plant.

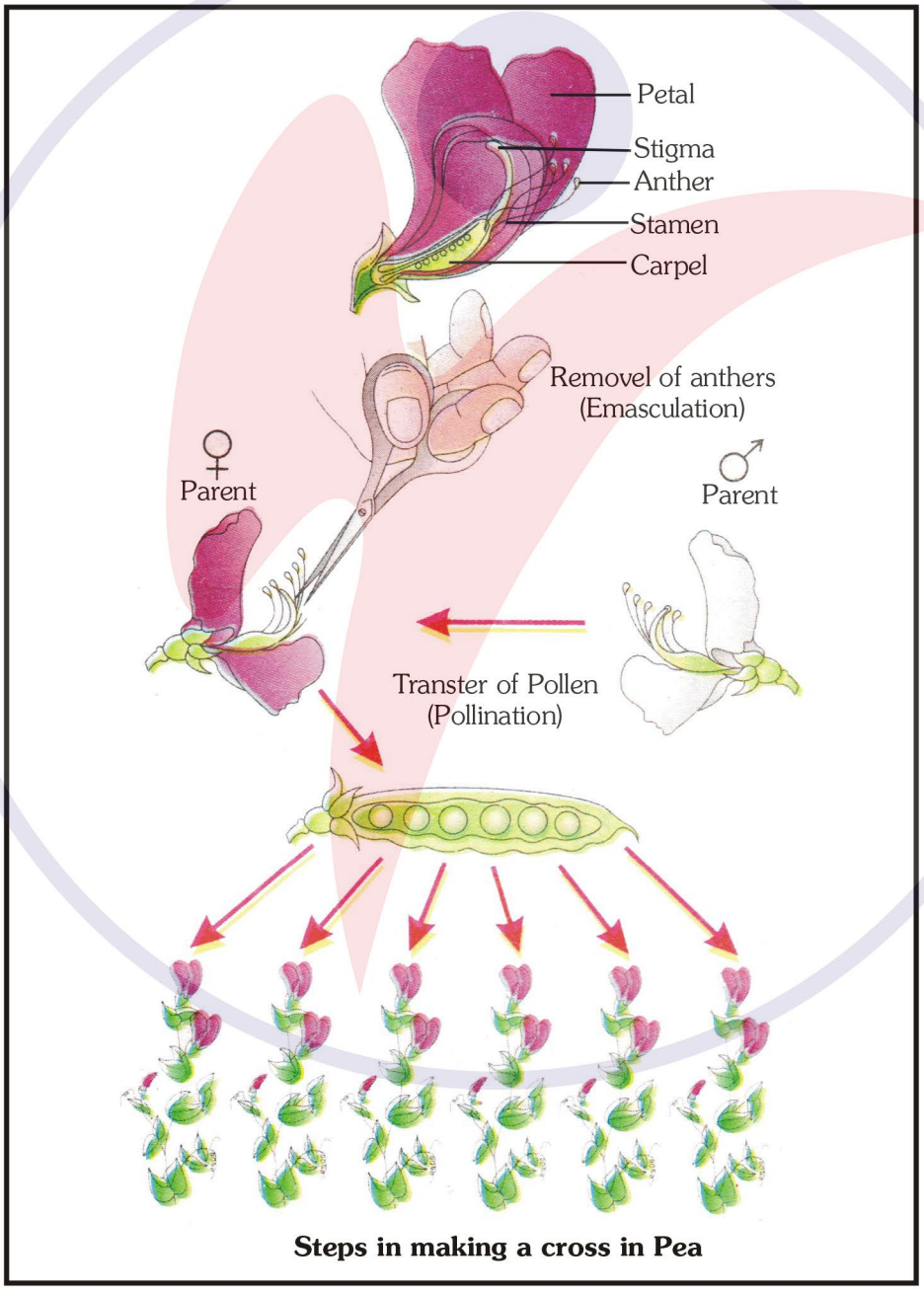
(iii) Mendel quantitatively analyse the inheritance of qualitative characters.

(iv) He maintained the statistical records of all the experiments.

(v) His experiment had large sampling size.

Mendel's work : Mendel studied 7 characters or 7 pairs of contrasting traits.

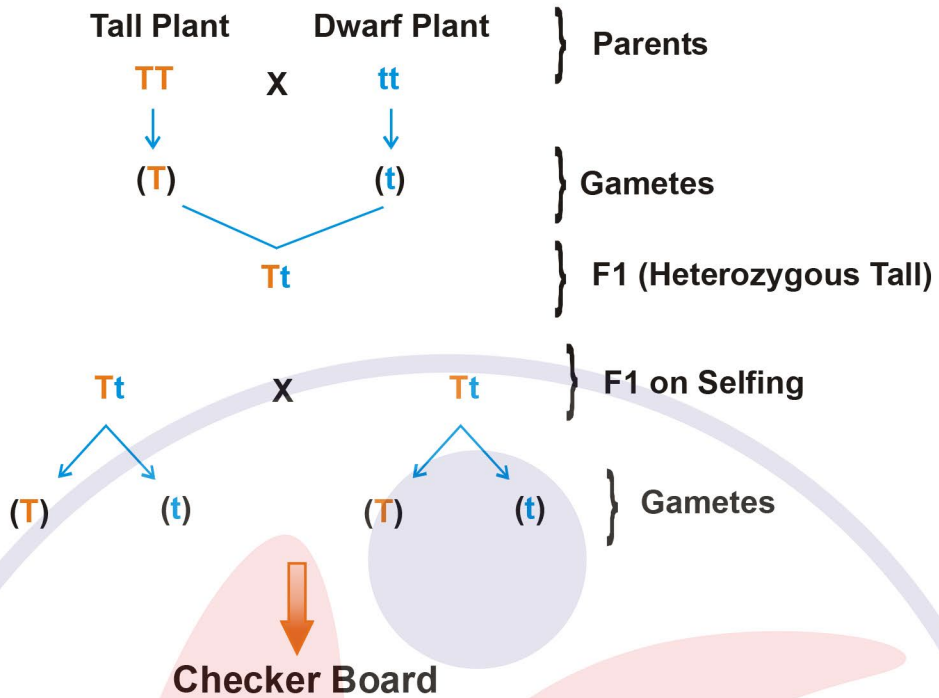
S.No.	Character	Ch. No.	Dominant	Recessive
1.	Length of plant	4th	tall	dwarf
2.	Flower position	4th	axial	terminal
3.	Shape of pod	4th	inflated	constricted
4.	Colour of pod	5th	green	yellow
5.	Shape of seed	7th	round	wrinkled
6.	Colour of colytedon	1st	yellow	green
7.	Colour of flower	1st	violet	white



Steps in making a cross in Pea

MONOHYBRID CROSS

When we consider the inheritance of one character at a time in a cross.



Checker Board

♀ \ ♂	(T)	(t)
(T)	TT *Homozygous Tall*	Tt *Heterozygous Tall*
(t)	Tt *Heterozygous Tall*	tt *Homozygous Dwarf*

Monohybrid Cross Ratio

Phenotypic ratio : 3 : 1 (3 Tall : 1 Dwarf)
Genotypic ratio : 1 : 2 : 1 (1 TT : 2 Tt : 1 tt)

Note :- Checker Board Method was used by **Reginald. C. Punnett**.

Conclusions (results) of Monohybrid Cross

Ist Conclusion (Postulate of paired factors) :

(i) Characters are controlled by discrete units called factors.
(ii) Factors occur in pairs.

IInd Conclusion (Postulate of Dominance):

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

Tall (TT) × Dwarf (tt)

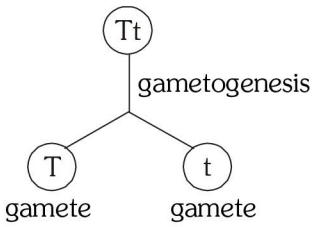
F_1-Generation (Tt) All tall

Law of dominance = Postulate of paired factors + Postulate of Dominance

There are two exceptions of law of dominance. [A] Incomplete dominance, [B] Co-dominance,

IIIrd Conclusion (Law of segregation):

During gamete formation ; the unit factors of a pair segregate randomly and transfer inside different gamete. Each gamete receives only one factor of a pair; so gametes are pure for a particular trait. It is known as conclusion of purity of gametes or segregation.



– The segregation is essential during the meiotic division in all sexually reproducing organisms. (Nondisjunction may be exception of this law).

Type of gamete /phenotypic category = 2^n

Type of genotype = 3^n

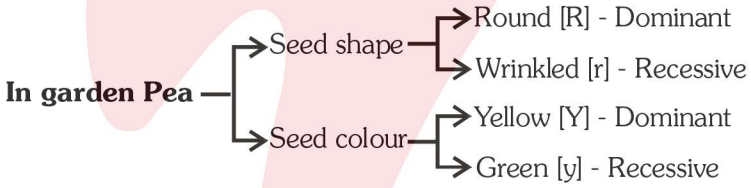
No. of zygote produced by selfing of a genotype = 4^n

n = No of hybrid character or heterozygous pair.

DIHYBRID CROSS

A cross in which study of inheritance of two pairs of contrasting traits.

Mendel selected traits for dihybrid cross for his experiment as follows :-



Mendel crossed, yellow and round seeded plants with green and wrinkled seeded plants.

All the plants in F_1–generation had yellow and round seeds.

When F_1 plants were self pollinated to produce four kinds of plants in F_2 generation such as yellow round, yellow–wrinkled, green round and green wrinkled, there were in the ratio of 9 : 3 : 3 : 1.

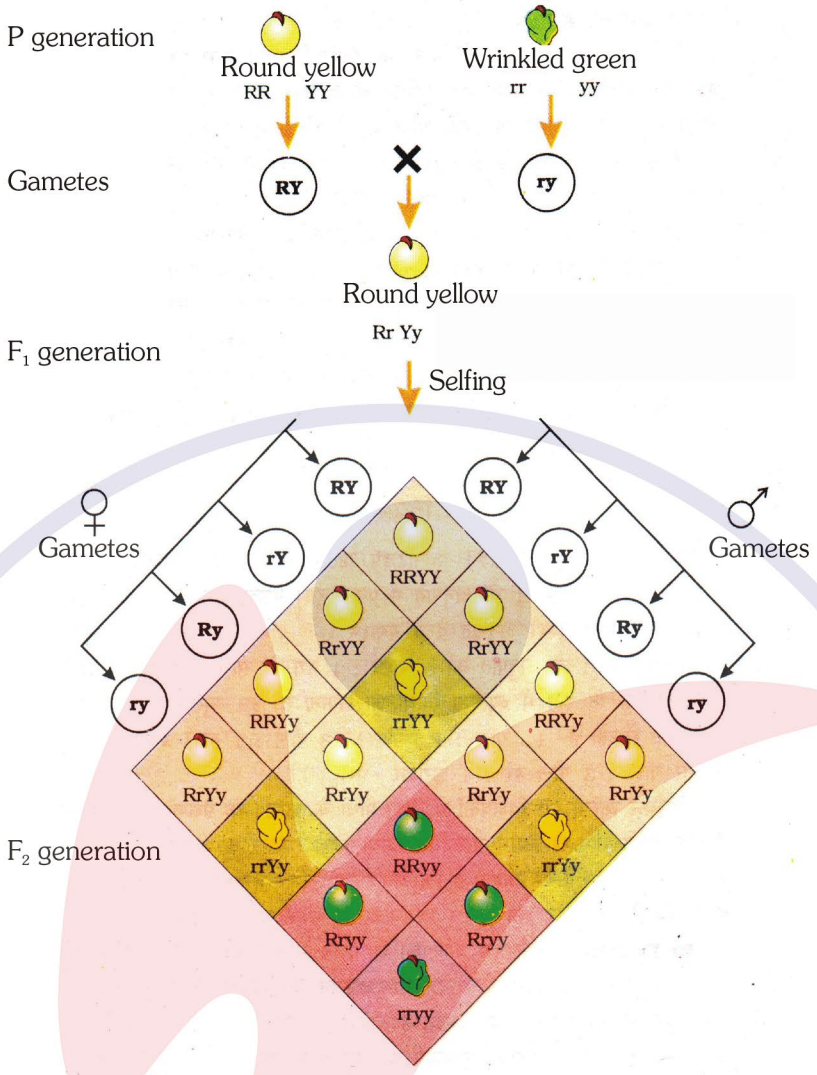
Phenotypic ratio : round yellow : round green : wrinkled yellow : wrinkled green
9 3 3 1

No. of Phenotype = 4

Genotypic Ratio :-

RRYY	RrYY	RRYy	RrYy	rrYY	rrYy	RRyy	Rryy	rryy
1	2	2	4	1	2	1	2	1

No. of Genotype = 9



Conclusion (Law of Independent Assortment):

The law states that "When two pairs of trait [2 different characters] are combined in a hybrid, segregation of one pair of character is independent of the other pair of character."

This is known as **Conclusion of Independent Assortment.** It is based on F_2 - generation of dihybrid cross.

Exception : Linkage

TEST CROSS

When F_1 progeny is crossed with recessive parent.

[A] Monohybrid Test Cross :-

F_1 progeny(hybrid) Recessive parent

T t × t t

↓

t

T	T t
t	t t

Monohybrid test cross ratio = 1 : 1

[B] Dihybrid Test Cross:-

F1 - dihybrid: T t R r × Recessive parent: t t r r

	RT	Tr	Rt	tr
tr	TtRr	Ttrr	ttRr	ttrr

Phenotypic Ratio = Genotypic Ratio = 1 : 1 : 1 : 1

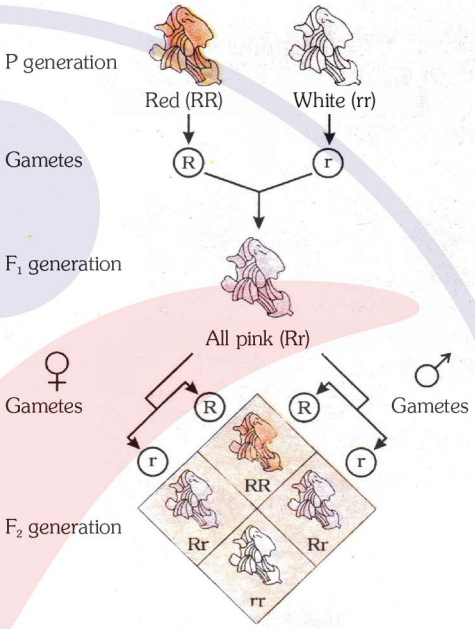
Conclusion:- (i) In test cross phenotypes and genotypes ratio are same.

(ii) **Test cross helps to find out the genotype of dominant individual.**

POST MENDELISM

[1] Incomplete dominance :-

F_1 phenotype did not resemble either of the two parents and was in between the two.



Example :-

(i) Flower colour in Mirabilis jalapa :

Phenotypic ratio : 1(red) : 2(pink) : 1(white)
Genotypic ratio : 1(RR) : 2(Rr) : 1(rr)

(ii) Flower colour in Antirrhinum majus (Snapdragon or Dog flower).

(iii) Size of starch grain in pea plant :

[2] Co-dominance :-

F_1 generation resembles both parent.

Examples :- (i) Co-dominance is seen in animals for coat colour.

when a black parent is crossed with white parent, a roan colour F_1 progeny is produced.

(ii) AB blood group inheritance (I^AI^B)

(iii) Carrier of Sickle cell anaemia (Hb^A Hb^S)

[3] Multiple allele :–

More than two alternative forms of same gene called as multiple allele. Multiple allele is formed due to mutation.

If n is the number of allele of a gene then number of different possible genotype = $\frac{n(n+1)}{2}$

Example of multiple allele :

ABO blood group → ABO blood groups are determined by three alleles – I^A, I^B, and I^O

I^A = dominant

I^B = dominant

I^O = recessive

Possible phenotypes - A, B, AB, O

Possible genotype number = $\frac{3(3+1)}{2} = 6$ genotype

Blood group	Genotype	Antigen or agglutinogen	Antibody or agglutinin
A	I^AI^A, I^AI^O	A	b
B	I^BI^B, I^BI^O	B	a
AB	I^AI^B	A & B	None
O	I^OI^O	none	a & b

[4] Pleiotropic gene :– Gene which controls more than one character is called pleiotropic gene. This gene shows multiple phenotypic effect.

For example :

(i) In Pea plant : Single gene influences → Seed shape, Size of starch grain

	Seed shape	Size of starch grain
BB	Round	Large
Bb	Round	Medium
bb	Wrinkel	Small

(ii) Sickle cell anaemia - (Autosomal recessive disorder/qualitative disorder)

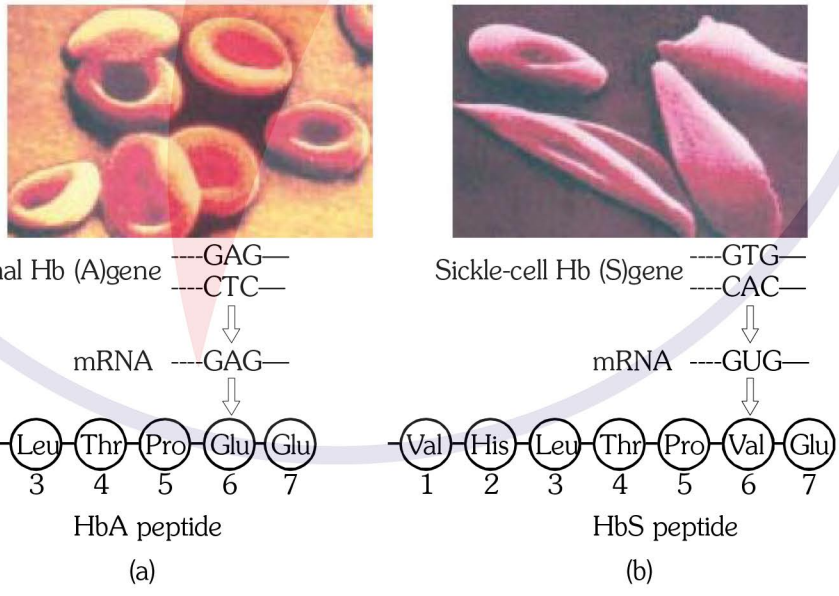
Possible genotype = 3

Hb^AHb^A = Normal

Hb^AHb^S = Carrier/Normal

Hb^SHb^S = S.C.A./Diseases [lethal]

The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG. The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.



Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin :

(a) From a normal individual; (b) From an individual with sickle-cell anaemia

POLYGENIC INHERITANCE

Inheritance of characters in which one character is controlled by many genes and intensity of character depends upon the number of dominant allele. Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment. In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive.

Examples :-

(i) Human height (regulated by 5 gene pairs).

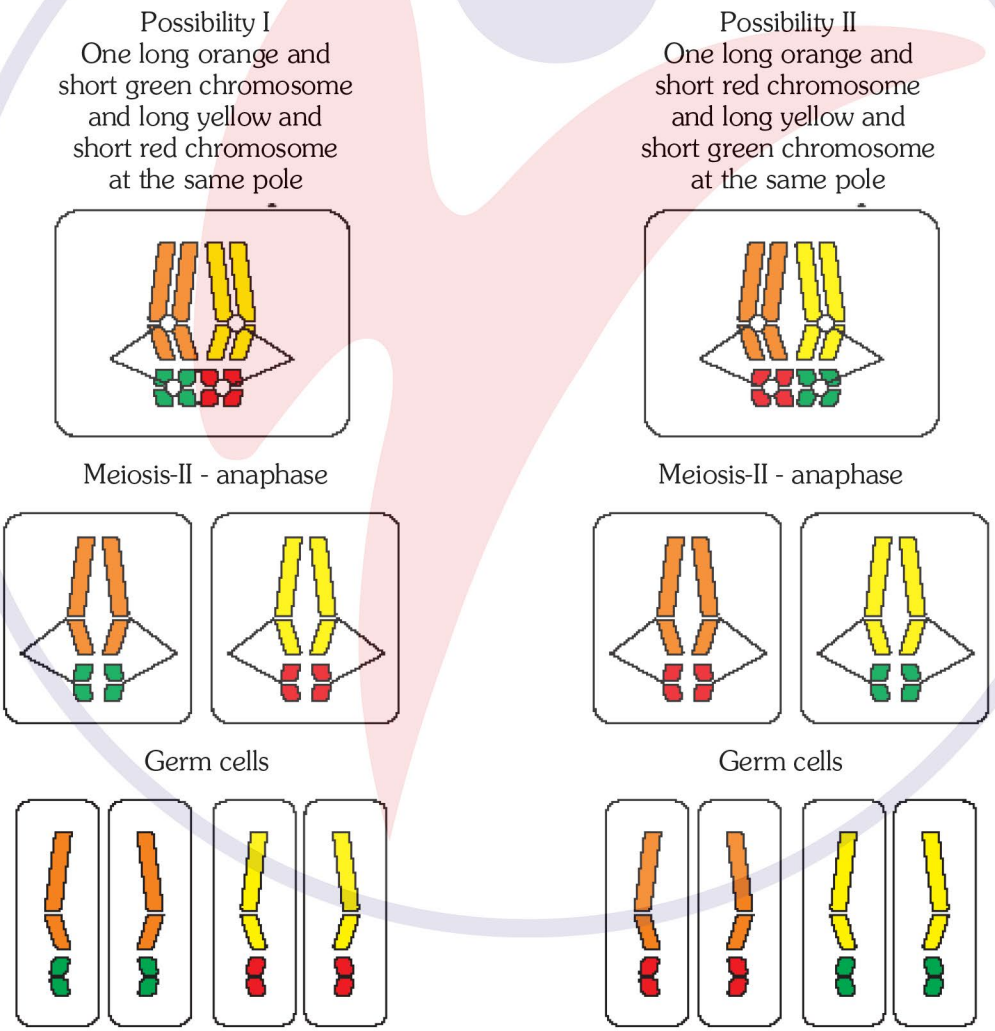
(ii) Human skin colour (regulated by 3 gene pairs).

When a Negro Black (AABBCC) phenotype is crossed with white (aabbcc) phenotype, intermediate phenotype produced in F_1 generation.

CHROMOSOMAL THEORY OF INHERITANCE

This theory was proposed by **Walter Sutton** and **Theodor Boveri** (1902).

Walter Sutton and Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws.



Independent assortment of chromosomes

A Comparison between the Behaviour of Chromosomes and Genes

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

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 as :-

1. They could be grown on simple synthetic medium in the laboratory.
2. They complete their life cycle in about two weeks,
3. A 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.

LINKAGE (By Morgan)

Linkage is the tendency of genes which are close together on a same chromosome to be inherited together.

Linked genes do not assort independently.

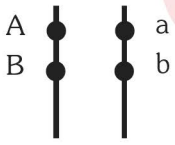
Genes present on same chromosome are called Linked genes.

Linked genes are non-allelic.

Crossing over breaks linkage.

Linkage and independent assortment can be represented in dihybrid plant, as –

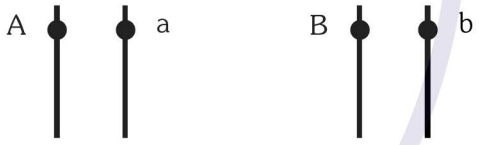
In case of linkage in dihybrid AaBb



It produces two types of gamete

AB : ab

In case of independent assortment in dihybrid AaBb



It produces four types of gamete

AB : ab : aB : Ab

Linkage Group : All the genes present on a chromosome collectively forms one linkage group.

Number of Linkage group = haploid no. of homologous chromosomes.

Types of Linkage :- There are two types of linkage –

Complete Linkage	Incomplete Linkage
• Rare type of linkage • Only parental combinations are found in gametes • No Recombination • Example: Male Drosophila	• Most common type of Linkage. • Parental as well as recombinant gametes are formed. • Recombination frequency less than or equal to 50%

• Distance can be identified by the incomplete linkage. It's unit is centi Morgan.

$$\text{Strength of linkage} \propto \frac{1}{\text{Distance b/w linked gene}} \propto \frac{1}{\text{Crossing Over}}$$

• **Genetic map/Linkage map/chromosome map -** In genetic map different genes are linearly arranged according to % of crossing over (∝ Distance) between them.

Morgan's student Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and 'mapped' their position on the chromosome.

LINKAGE : RESULTS OF TWO DIHYBRID CROSSES CONDUCTED BY MORGAN

Cross A shows crossing between gene y (body colour) and w (eye colour)

Cross B shows crossing between genes w (eye colour) and m (wings size).

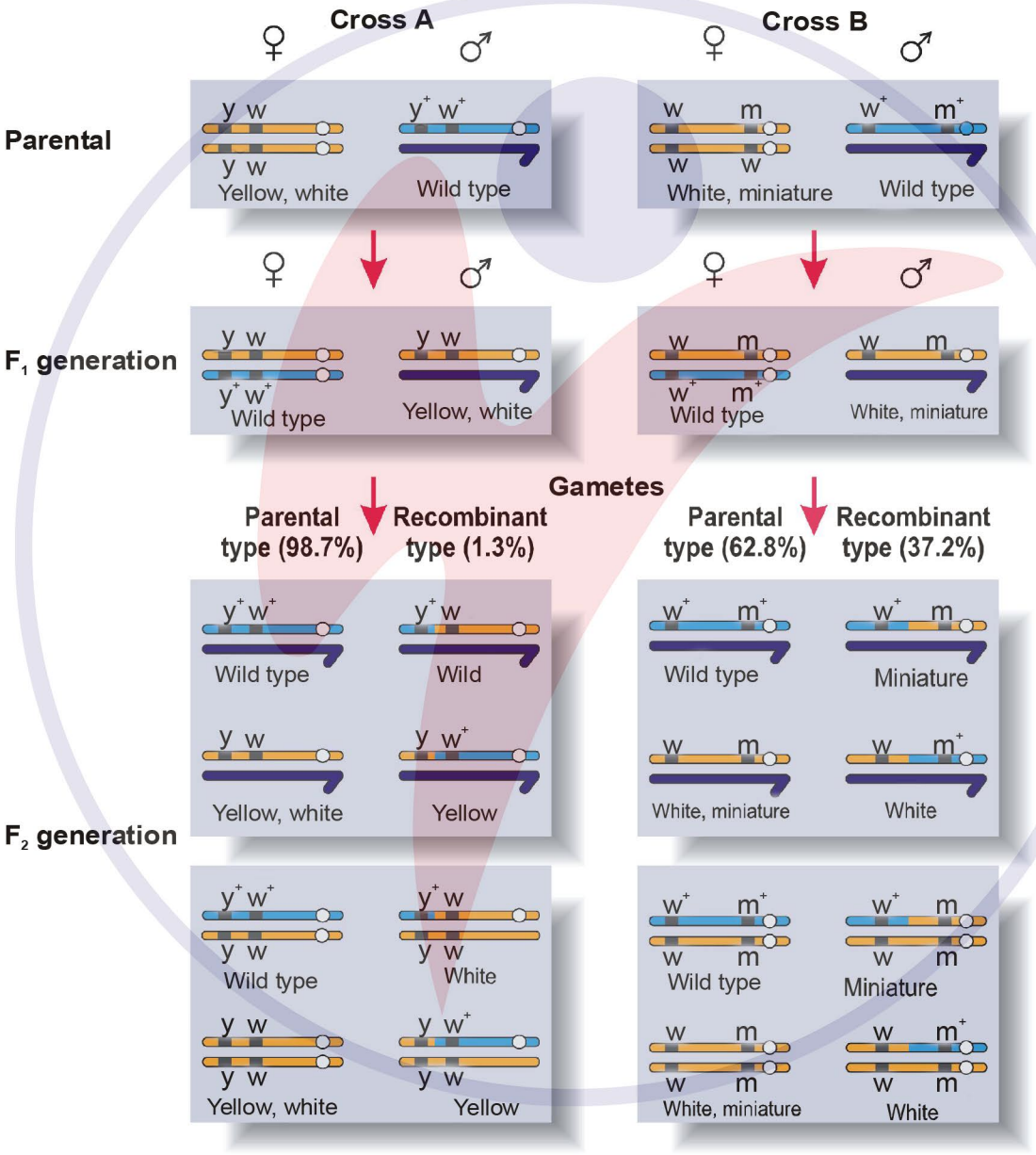


Figure : Results of two dihybrid crosses conducted by Morgan.Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alletes are represented with (+) sign in superscript. Note : The strength of linkage between y and w is higher than w and m.

SEX LINKAGE

When the genes are present on sex-chromosome is termed as sex linked gene and such phenomenon is known as sex-linkage.

Types of sex linkage :-

1. **X-linkage.**

Genes of somatic characters are found on x-chromosome.

Example of X-linkage :-

[i] Eye colour in Drosophila :- Eye colour in Drosophila is controlled by a X–linked gene.

[ii] Haemophilia (bleeder's disease/royal's disease) :-

The gene of haemophilia is recessive and x-linked lethal gene.

On the basis of x-linked, following types of genotype are found.

X^h X = Carrier female

X^hX^h = Affected female

X^hY = Affected male.

But, X^hX^h type of female dies during embryo stage because in homozygous condition, this gene becomes lethal and causes death.

[iii] Colour Blindness (X-linked recessive) :- Due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour.

[iv] Diabetes insipidus (recessive).

[v] Pesudoricketes (Dominant)

2. **Y- linkage -** The genes of somatic characters are located on Y- chromosome. The inheritance of such type of character is only through the males. Such type of character is called Holandric character. These characters found only in male.

Example :- (i) Gene which forms TDF /sry-gene

(ii) Hypertrichosis (excessive hair on ear pinna.)

SEX DETERMINATION

Establishment of sex through differential development in an individual at an early stage of life, is called sex determination.

Mechanism of sex determination :

[I] Allosomic determination of sex –

Method	Female	Male	Examples
XX-XY	Homogamatic	Heterogamatic	Human Drosophila
ZW-ZZ	Heterogamatic	Homogamatic	Birds, Reptiles
XX-XO	Homogamatic	Heterogamatic	Grasshopper, Cockroach

X- Chromosome discovered by ***"Henking"*** and called 'x-body'.

[II] Haploid - diploid mechanism (Sex determination in Honey Bee) –

Diploid (two sets) → Female [32-chromosomes]

Haploid (One set) → Male [16-chromosomes]

In honey bee, male individual (Drone) develops from unfertilized eggs (Haploid). Male is always parthenogenote. Queen and worker bees develop from diploid eggs i.e. fertilized egg.

[III] Cytological basis of sex Identification (Barr body technique) **–**

Interphasic nucleus of human female contains two X- chromosomes. Out of two, one X- chromosome becomes heterochromatin and other X- chromosome is euchromatin. By staining X- heterochromatin, it appears as a dense body which is called Barr body.

No. of Barr body ⇒ (No. of X chromosomes – 1)

HUMAN GENETICS

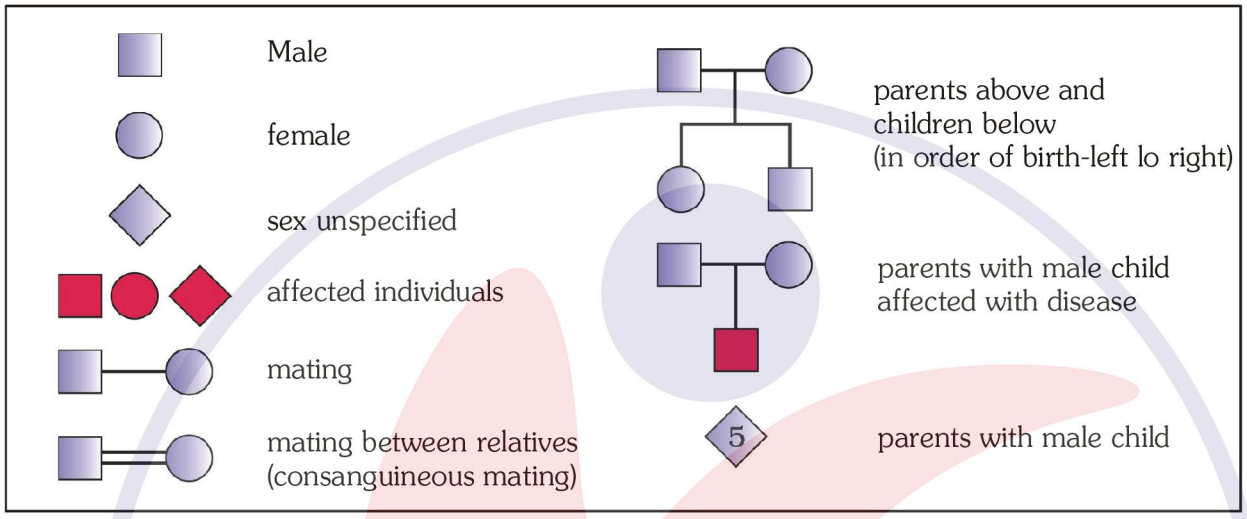
The study (analysis) of genetic characters and aspects like genetic improvements among humans are included in **human genetics.**

The study and analysis of human genetics is performed by many methods like pedigree analysis, statistical analysis and human karyotyping.

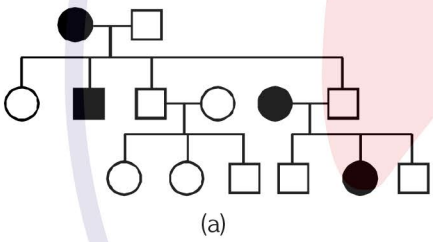
Pedigree Analysis

Study of ancestoral history of man of transmission of genetic characters from one generation to next, is pedigree analysis.

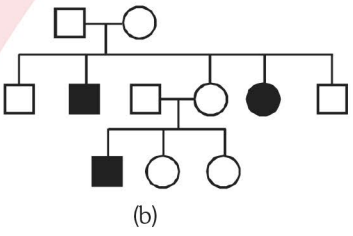
To study and analyse them a pedigree of genetic facts/data and following symbols are used.



Examples :-



(a) Autosomal dominant trait (Mytotonic Dystrophy)



(b) Autosomal recessive trait (Sickle Cell Anaemia)

Special Note :-

Autosomal Disorder

Autosomal Dominant
AA ⇒ Affected
Aa ⇒ Affected
aa ⇒ Normal
Ex. Mytonic dystrophy
Polydactyly

Autosomal recessive
AA ⇒ Normal
Aa ⇒ Normal
aa ⇒ Affected
Ex. Albinism
Phenylketonuria
Alkeptonuria
Sickel cell Anaemia
Thalassemia
Cystic fibrosis