Chapter 22

VARIATION AND GENETICS

IMPORTANT TERMS

Genetics:

It is the branch of biology which deals with the hereditary characteristics of an individual and their transmission from parent to offspring.

Tralt or Character:

A character that is controlled by a pair of genes is called a trait or a character.

Factor:

Gene which is responsible for a particular trait is also called a factor.

Dominant:

The trait that appears in the offspring of two parents is called dominant trait. The gene controlling this trait is also said to be dominant.

Recessive:

The trait or gene that is **suppressed** or **masked** in F1 generation of two ture breeding organisms is called recessive e.g., the gene of dwarfness **(t)** is recessive.

Phenotype:

Phenotype is the form of appearance of a trait e.g., **color, from, size behavior** etc.

Genotype:

Genotype is the genetic complement i.e. genes in an individual for a particular trait such as TT, Tt, tt etc. (It refers the genetic makeup of a trait of the individual)

Homozygous:

When both the alleles of gene pair are same then they are heterozygous such as **Tt, Rr** are heterozygous.

Hybrid:

A heterozygous individual is also called **hybrid**.

Monohybrid and dihybrid:

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Monohybrids are the offspring of parents that differed in **one character** only, while dihybrids are offspring of parents that differ in one contrasting pair of trait.

Genes:

The smallest part of DNA is called genes which is a **basic unit** of biological information.

Explanation:

The biological information in **DNA** is stored and **coded** in the sequence of its **bases**.

Locus:

The position on a chromosome where a gene is located is often referred to as locus. As genes are passed from one generation to another so it can pass the same characters from parents to offsprings and can thus produce inherited resemblances but they are also responsible for producing many distinctive variations as well in next generation. Variations are produced due to the reshuffling or mutation of the genes.

Jumping Genes:

Jumping genes do not settle peacefully on their loci, they keep on moving or jumping on different loci on the same chromosome or other chromosomes.

Alleles:

A single gene may have alternative form which is called allele, or partners of gene pair are called allele.

Explanation:

Each allele of a gene pair occupies the same gene locus on its respective **homologue**. Both alleles on one locus may be identical or different from each other.

Genotype:

The specific alleles or genes contained in a cell are called **genotype**.

Phenotype:

The physical trait that occurs as a result of specific genotype is referred as ***phenotype***.

Example:

The concept of **phenotype** and **genotype** can be understood by taking an example of flower colour. Flower colour is a trait and red and **white** are its two phenotypes. Each

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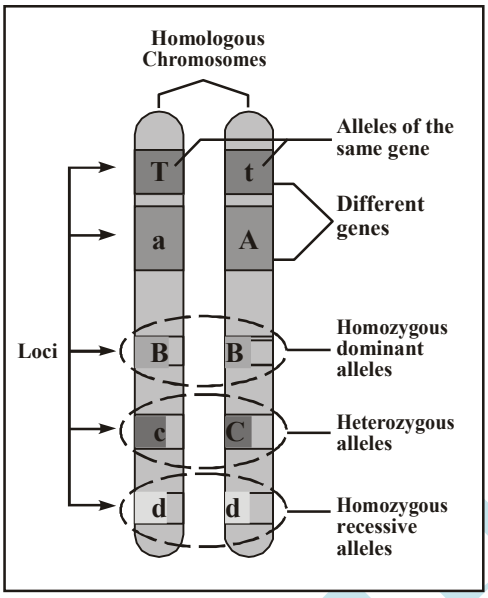
form of expression is determined by different allele of colour gene. **Allele R** is the determiner' for redness while 'r' is determiner for whiteness.



Allelic pairs on a homologous pair of chromosomes

Gene Pool:

Definition:

The group of individuals of same species that interbreed sexually and live in same place are called **population**. The genes distributed among all individuals of a population are collectively called a **gene pool**. It is the total genetic information encoded in the total genes in a breeding population existing at a given time.

Example of bean bag genetics:

To have clear understanding we can have the concept of **bean bag genetics**. The alleles are like **beans** in a beanbag. The entire **bean bag** full of beans is the **gene pool** of population. In the beanbag approach we can imagine the entire gene pool comprising all the alleles for all the different traits at once, or we can just focus on some **subset**, such as all the alleles for single trait.

A sample population of 100 **diploid plants**, some of which bear **red flowers** others bearing **white flowers** has sum total of **200** of all different alleles **(R or r)** for flower colour trait as its gene pool.

Conclusion:

Thus it is found that all alleles in a gene pool are dispersed in individuals and determine the genotypes present in population at any one time.

Importance of Pea Plant:

Gregor Johann Mendel:

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Gregon Johann Mendal (1822-1884) is considered as the **father of genetics**. He was a **priest** but he was interested in **plant breeding**. He worked on the **pea plant** and formulated the two principals: **Law of segregation** and law of **independent assortment**.

Choice of Pea Plant:

Mendel's work was on pea plant **Pisum sativum** and grow it in his **monastery garden** for **eleven years**. He chose this plant for **many reasons:**

1. It was **easy to grow**.
2. **Flowers** of pea plant were **hermaphrodite**.
3. It was **self fertilizing plant** but it can be cross fertilized.
4. The time gap between the generations was short, Mendel could produce many generations of pea within a short time.
5. Pea plants had many sharply distinct traits.
6. Each trait had two clear alternative forms or varieties, that differed from each other in very pronounced (sharp) ways e.g. seed colour could be **yellow** or **green**. Mendel called them **contrasting pair of trait.**

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Plant Height	Tall (6-7 feet) 	Short (9-18 inches) 
Flower Colour	Purple 	White 
Flower Position	At leaf junctions (axial) 	At tips of branches (terminal) 
Pod Colour	Green 	Yellow 
Pod Shape	Inflated 	Constricted 
Seed Colour	Yellow 	Green 
Seed Shape	Round 	Wrinkled 

Seven traits of garden pea studied by Mendel.

LAW OF SEGREGATION

Statement:

This law state that co-existing alleles for each trait in an individual segregate from each other so that each gamete receives only one of the two alleles. Alleles unite at random fertilization of gametes.

Explanation:

He first of all produced **true breeding** strains of each trait.

True breeding strains are those that consistently, generation after generation, yield offspring with the same traits e.g. a true breeding round seed plant produces only round seeds.

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Similarly a true breeding wrinkled seed plant produce only wrinkled seeds. After establishing, 14 pure breeding lines of **seven character** he cross fertilized plants that differed in one character only. The offspring of such a cross were called **monohybrids**. He cross fertilized true breeding round seed male plant with true breeding **wrinkled** seed **female plant**.

F_1 Generation and Principle of Dominance:

He called it **first parental generation (P_1).** The offspring produced from the cross of P1 generation are called **F_1 or first filial generation.**

With the cross of round and wrinkled all the offsprings of **F_1** were **round**. These results were same with one of their parent round but no wrinkled phenotype appeared. Mendel termed the trait that appeared in F_1 generation the **dominant trait** and he described the one that had failed to appear as **recessive trait**.

F_2 Generation:

He allowed the F_1 plants to self fertilize to produce **F_2 second filial generation**.

Mendel found the following result.

1/3 were green and

3/4 were round

He repeated the experiment with other pea strains and got the same ratio of 3 : I in offspring of monohybrid crosses for all seven contrasting pair of traits.

F_3 Generation:

Mendel performed more experiments to get **F_3 (third filial generation)** by self fertilizing F_2 plants. He noted that 1/3 of F_2 round produced only round, while 2/3 of F_2 round produced both round and wrinkled in 3 : I ratio but F_2 wrinkled produced only wrinkled. He concluded that 1/3 of F_2 rounds were true breeding like F_1 round and 2/3 of F_2 round were **monohybrids** like F_1 round.

Homozygous and Heterozygous:

From his experiments Mendel realized that there are two kinds of round seeds. One is of **true breeding** kind which will produce the plants like that of their parents. The other resembles **F_1** generation which produces the plants containing both **round and wrinkled** genotypes. Thus the true breeding round is called **homozygous** and other kind **heterozygous**.

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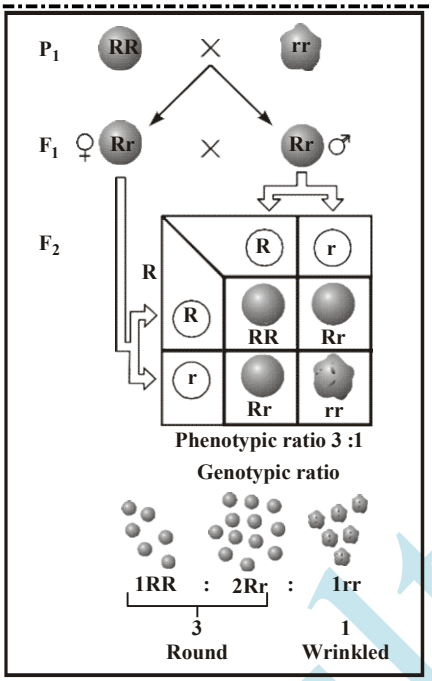


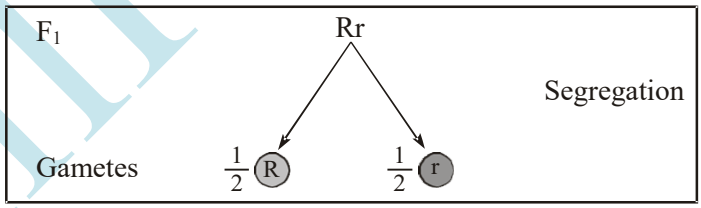
Mendel's cross to study single trait inheritance in pea

When both the alleles of a gene pair in an organism are same, the organism is **homozygous** for that gene pair. An individual with homozygous genotype is homozygote. **Heterozygous** means that organism bears two different alleles of gene pair.

Mathematical Representation:

Punnet square indicates that 1/4 of F_2 progeny would have been RR (homozygous round).

$\frac{1}{4} + \frac{1}{4} = \frac{1}{2}$ Rr (heterozygous round and $\frac{1}{4}$ rr (wrinkled).



Segregation of alleles during gamete formation

Mendel's Interpretation:

Mendel inferred from his experiments that:

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(i) A trait is produced by at least a pair of factors. That pair may be **homozygous** or **heterozygous**. Mendel found that **factors** of a pair separated from each other during **gamete formation**.

(ii) Each gamete receives one of a pair of factors so half of gametes got one allele and other half carried other allele.

(iii) On fertilization offspring receives only one factor from each parent. It one parent's factors are **heterozygous** the offspring has an equal chance of receiving either factor from that parent.

(iv) Where dominance is found, the dominant factor will be expressed over the **recessive**, the recessive will be expressed only when two recessive factors come together in offspring.

TEST CROSS AND ITS IMPORTANCE

Test Cross:

Definition:

Test cross in a mating in which an individual showing a dominant phenotype is crossed with an individual showing its recessive phenotype.

Importance:

This cross finds out the **homozygous** or **heterozygous** nature of **genotype.**

Explanation:

Mendel devised a cross to test the genotype of an individual showing a dominant phenotype. The seed having the **phenotype round** may be **homozygous (RR)** or **heterozygous (Rr).**

Case I:

It the seed is **homozygous round (RR)** it will grow into a pea plant that forms all gametes with only **'R' allele.** Fertilization will result in **100% round seed progeny.**

Result:

All round seed progeny.

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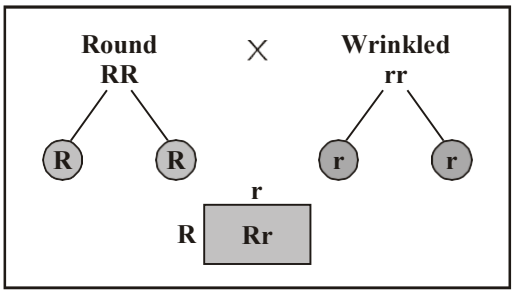
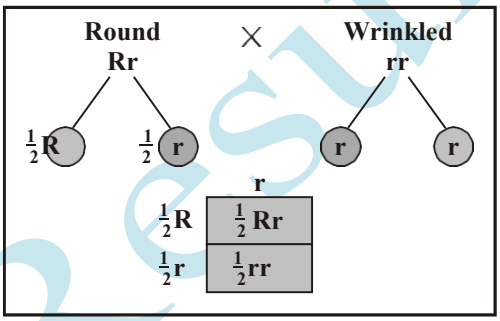
The tested phenotypically dominant individual is homozygous.



Case II:

If the seed is heterozygous round (Rr), it will grown into plant that forms half the gametes with 'R' and half with 'r' allele. Wrinkled seed plant will form only 'r' type of gametes. Fertilization will result into 50% round and 50% wrinkled seed progeny is a convincing proof for heterozygous nature of the round parent.



Result:

$\frac{1}{2}$ round seed and

$\frac{1}{2}$ wrinkled seed progeny. The tested phenotypically dominant individual is **heterozygous.**

Law of Independent Assortment:

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Statement:

When two contrasting pairs of traits are followed together in the same cross, their alleles assort independently into gametes.

Explanation:

After studying inheritance of two traits simultaneously e.g. **seed shape** and **seed colour**. He wanted to see if there was any relationship between the inheritance of one gene and inheritance of other gene.

Dihybrid Cross:

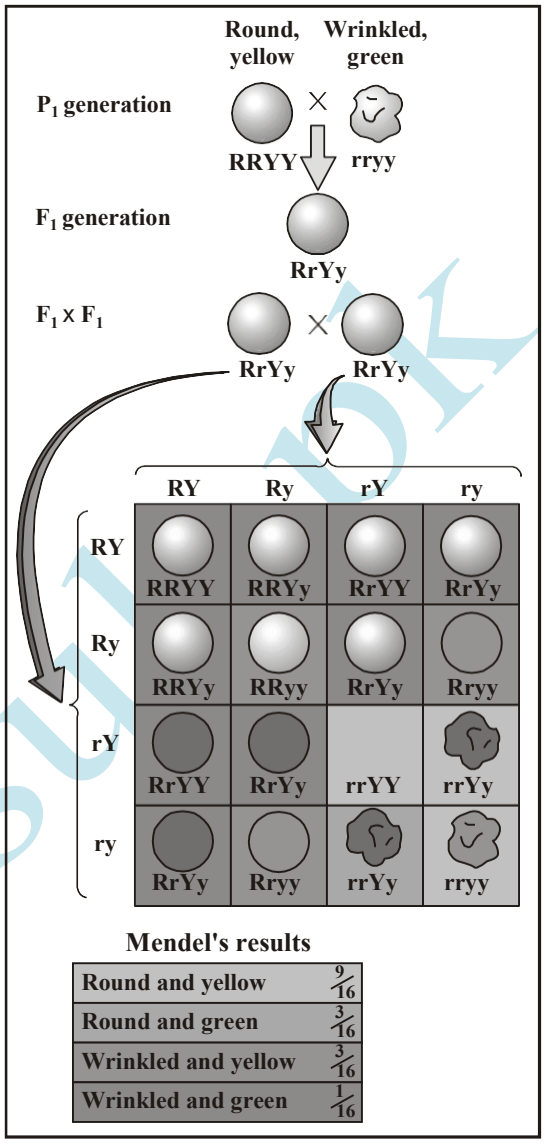
He crossed a **true breeding round yellow plant** (i.e., pea plant having round seeds with yellow colour) with another true breeding strain that bore **wrinkled green peas.** The F_1 offspring were all tound and yellow due its dominance. The symbols given to them were;

In which **R and r** will be symbols for round and **wrinkled** respectively and **Y and y** will be symbols for two alleles of **yellow** or **green**. Mendel got the **F_2** generation by crossing **$F_1 \times F_1$.**

(Rr Yy × Rr Yy)

Round yellow	9
Round green	3
Wrinkled yellow	3
Wrinkled green	1

F2 progeny were not only in two parental combinations i.e. **round yellow (9)** and



Dihybrid cross produces parental as well as recombinant types.

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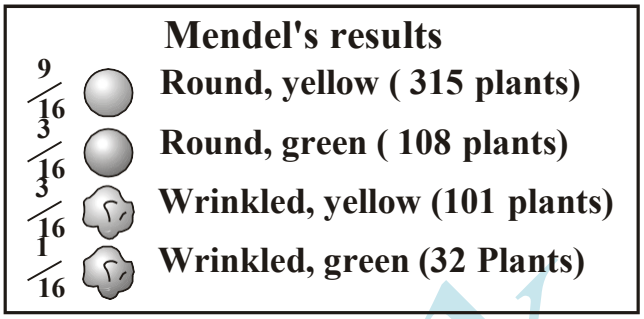
wrinkled green (1) but also in new phenotypic combination i.e. **round green (3)** and **wrinkled yellow (3)**. A phenotypic ratio of 9:3:3:1 appeared in F2. This new Phenotype indicates that some sort of shuffling of alleles had occurred during gamete formation.



Mendel's Conclusion:

He concluded that alleles for **seed shape** and colour were not bound to remain in parental combinations for ever i.e. **R** with **Y** and **r** with **y**. They can **assort independently**. **R** could go with Y or y in any gamete with **equal chance**. Similarly **r** could go with **y** or **Y** any gamete with equal probability. Four types of gametes i.e. **Rr, Ry, rY** and **ry** were formed in equal number in perfect ratio of **1:1:1:1**. When these gametes **randomly** fertilized each other, **9:3:3: I phenotypic** ratio of found in F_2 progeny.

Results Derived from Mendel's Conclusion:

The inheritance of each pair of differing traits is **independent** of any pair of **traits** or by correlating it with meiosis. The way in which a pair of alleles on one set of homologous chromosomes **segregates** during meiosis has no effect on the way a pair of alleles on another set of homologous chromosome segregates.

Genes are located at specific **loci** on chromosomes. In **dependant assortment** of genes depends upon independent assortment of their chromosomes. All the genes present on a homologous pair of chromosomes are **linked** to each other in the form of **linkage group**. These cannot assort independently. Only those contrasting pairs of traits can assort independently whose alleles are riding **non homologous chromosomes.**

PROBABILITY:

Definition:

It is a chance of an event to occur.

Example:

To understand this concept we have to study the inheritance of a **seed shape** which is an **independent** event. In F_2 offspring of a seed to be round is ¾ or it to be wrinkled is ¼.

Inheritance of seed colour is another separate event. The independent chance in F_2 of a monohybrid cross for a seed to be yellow is ¾ or it to be green is ¼.

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Product Rule:

Event No. 1	Event No. 2	Both events at a time
Seed shape	**Seed colour**	**Seed shape and colour**
Independent probability to be:	Independent probability to be:	Joint probability of being:
Round = ¾ Round = ¾ Wrinkled = ¼ Wrinkled = ¼	yellow = ¾ green = ¼ yellow = ¾ green = ¼	Round yellow = ¾ x ¾ = 9/16 Round green = ¾ x ¼ = 3/16 Wrinkled yellow = ¼ x ¾ = 3/16 Wrinkled green = ¼ x ¼ = 1/16

When two independent events are occurring **simultaneously** like in **dihybrid** cross, the ratio of each joint phenotypic combination can be obtained by **multiplying** the **probabilities** of individual phenotypes. It is **product rule**. The joint probability that both of the independent events will occur simultaneously, is equal to **product** of individual probabilities of each event.

Dominance Relationships:

Definition:

Dominance relationship are the various in which the two alleles at one gene locus can affect the phenotype.

There are four types of dominance relations among alleles.

1. Complete dominance
2. Incomplete dominance
3. Co-dominance
3. Over-dominance

1. Complete Dominance:

In this type of relationship the information from one allele will **completely be ignored** while other allele will completely determines the phenotype.

Example:

In the example of **round** and **wrinkled** seeds one allele **R** is completely **dominant** over the r, presence of recessive allele is functionally hidden, so heterozygote **(Rr)** has same round phenotypes as **(RR)** homozygote. The contrasting pairs of allele for all **seven characters** chosen by Mendel showed complete dominance.

2. Incomplete Dominance:

It is also called **partial dominance** where the heterozygote appears to be a **blend** of two **homozygotes**.

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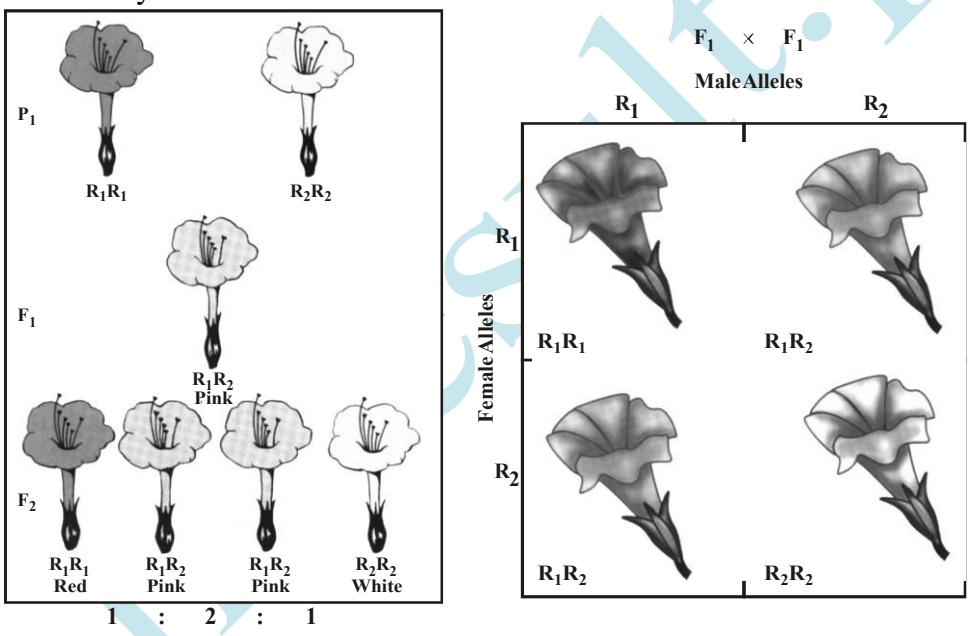
Example:

A calssic example of incomplete dominance is found in flowering plant named **4 O'clock**. In **1899 Carl Correns** worked on these flowers. When he crossed a true breeding **red flowered plant** with true breeding **white flowered 4'O clock**. F1 hybrids has **pink flowers**. The new type of phenotype produced has a shade **intermediate** between those of the parents due to an **intermediate amount** of pigment in petals. To find the more clear results he self fertilized F1 pink flowers. The F2 showed all three phenotypes of flowers in the ratio of **I red : 2 pink 2 white**. Red was **homozygous** for red alleles and white was homozygous for white alleles. But when allele for each other and thus due to incomplete dominance of each other's effect **pink colour** was produced.

Explanation:

Incomplete dominance is always **capital** letters for both alleles e.g. **R_1** is **Red** and **R_2** for **white** and small letter distinction is not used. When this cross is performed on punnet square it shows that both phenotypic and genotypic ratios are same i.e. **I : 2 : 1**. There is absolutely no need of test cross.



Incomplete dominance in 40' clock

3. Co-Dominance:

Co-dominance is the equal expression of both alleles that result in a **mixed phenotype**. This kind of co-expression is most easily seen in some biochemical phenotypes like blood types heterozygote. The phenotype in this condition is completely different in quality from those of two homozygotes. The phenotype is not intermediate. Each allele of gene pair is associated with different substance.

Allele A ⟶ Substance x

Allele A2 —Produces→ Substance y

Co-dominance occurs when both the alleles express independently in heterozygote **(A1A2)** and from their respective products **X** and **Y**. the co-dominant heterozygote would have both substances at same time.

MN Blood Type OR Blood group system:

Human blood groups can be of many types, e.g. **ABO, MN, MNSs, Rh** etc.

Landsteiner and **Levine** discovered **MN blood types** in man on basis of specific **antigens** present produce the specific **antibodies**.

Basically there are two types of antigens. **M antigen** which is produced, by gene L^M for M phenotype and N antigen produced by L^N for N phenotype. While MN antigens are produced simultaneously by allele L^M and L^N for MN phenotype.

Phenotype	Genotype	Antigens on RBC
M	$L^M L^M$	M
N	$L^N L^N$	N
MN	$L^M L^N$	M and N

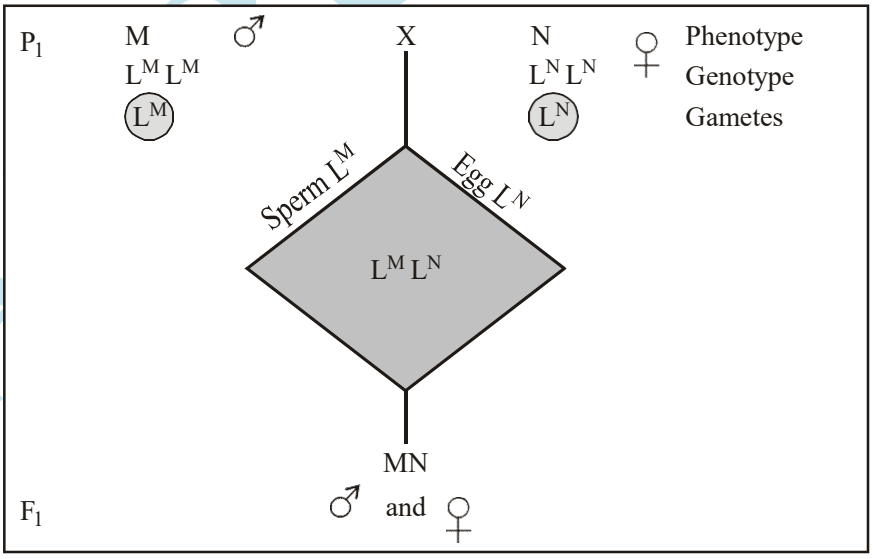
If a man of **M** blood group marries a woman of **N** blood group all their children will have **MN blood group**.

4. Over Dominance:

In this type of relationship the dominant heterozygote exceeds in quantity the phenotypic expression of both **homozygotes.**

Example:

The example of over dominance in presence of **fluorescence pigments** in eyes of ***Drosophila***. In fruit fly ***Drosophila*** the heterozygote has genotype (W^+ / W). It has more quantity of **fluorescent pigments** in **eyes** than **wild** (W^+ / W^+) or white eye (w/w) homozygote.



MULTIPLE ALLELES

Definition:

All the altered alternate forms of a gene, whose number is more than two, are called multiple alleles.

Some genes may have as many as **300 Alleles.**

ABO Blood Group System in Man:

It is the first discovered as multiple allelic system in man. A person having **antigen A** has **blood group A**; a person having **antigen B** has **blood group B**; a person having both the **antigens A** and **B** has **blood group AB;** but a person having **neither antigen A** nor **B** would have **blood group 0.**

Genetic Basis of ABO Blood group system:

Bernstein explained the genetic basis of **ABO** system in **1925**. This blood group system is encoded by a single polymorphic gene I and I is located on **chromosome 9**. It has three multiple alleles $\mathbf{I^A}$, $\mathbf{I^B}$ and **i**.

Blood group	Genotype	Antigen on RBC	Antibody in Serum
A	$I^A I^A$ or I^A i	A	B
B	$I^B I^B$ or I^B i	B	A
AB	IA IB	AB	Nil
O	ii	Nil	AB

Allele $\mathbf{I^A}$ specifies production of **antigen A** and **allele $\mathbf{I^B}$** specifies production of **antigen B** but **allele i** does not specify any antigen. Allele $\mathbf{I^A}$ and $\mathbf{I^B}$ are **co-dominant** to each other, because each expresses equally in $\mathbf{I^AI^B}$ heterozygote to produce **AB** phenotype. But allele **i** is recessive to both $\mathbf{I^A}$ and $\mathbf{I^B}$. Therefore $\mathbf{I^AI^A}$ or $\mathbf{I^Ai}$ genotypes will produce phenotype **A.** Similarly $\mathbf{I^BI^B}$ or $\mathbf{I^Bi}$ produces phenotype **B**. The homozygous ii will produce phenotype **O.**

Expression of the Alleles:

The blood group alleles start their expression at **early embryonic stage** and keep on expressing themselves **till death**. Therefore blood group phenotype of a person **never changes.**

Antiserum:

The blood serum containing **antibodies** is called **antiserum**. Anti A and anti B antibodies appear in plasma during the first few months after birth. They naturally occurring in the absence of corresponding antigen.

The blood serum of **A phenotype** contains **anti-B antibodies**. They will agglutinate or clump any RBC which have B antigens on them.

B phenotype contains anti **A antibodies** in the serum and **agglutinate** any RBC with antigen A.

AB has **neither anti A nor B antibodies** in the serum.

The serum of **0 blood** type contain **both anti A and anti B antibodies.**

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Blood Transfusion:

Any blood **transfusion** (process of transfer is ideally safe if it does not cause **agglutination** in the recipient (person who take blood). Agglutination leads to serious results **clumped** cells cannot pass through fine **capillaries** and may lead to embulus formation. If incompatible blood is transfused dangerous hemolytic reaction occurs. Either the antibodies of the recipient destroy the RBC of donor or the antibodies of the donor **hemolyze** the RBC of the recipient.

Safe Blood Transfusion:

Blood group A can be transfused only into A and AB recipients because they do not have **anti A antibodies.**

Blood group B can be transfused only into B and AB recipients as they do not have **anti B antibodies.**

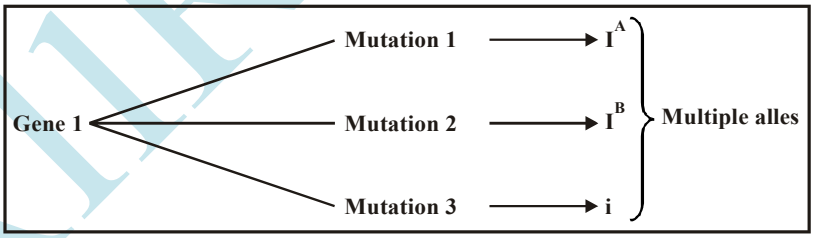
AB blood can be transfused only into AB recipients because they have neither **anti A, nor anti B antibodies.**

O blood can has neither A nor B antigens but it does have anti A and anti B anti bodies. An O recipient can only be given transfusion from a donor 0. Phenotype 0 can also be used as donor for small transfusion to A, B and AB recipients blood stream. O blood group individuals are called **universal donors. AB** blood group individuals are called **universal recipients** because they can receive transfusion of blood from any of the four blood groups.

Genetic Analysis of blood Groups:

Genetic analysis or the basis of blood groups help in solving cases of **Disputed parentage.** It can only be used to prove that an individual is not the parent of a particular child, e.g. a child of **AB** phenotype **(I^A I^B)** cannot be the child of a parent of phenotype **(O-ii).**

Similarly a man of **B** phenotype cannot be father of a blood type **A child,** whose mother is of phenotype **O.** His father could either be **A or AB** phenotype.



RH BLOOD GROUP SYSTEM

Rh Factor:

ABO blood type is further differentiated by a (+) or (---) sign. This **positive** or **negative sign** refers to the presence or absence of another blood group system antigen called **Rh factor.** Rh blood group system is defined on the basis of Rh factor present on the surface of **RBC.** This system is named Rh after **Rhesus monkey,** because its antigen was first discovered in it by **Landsteiner in 1930s.**

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Genes of Rh Blood:

Rh blood group system is encoded by three genes **C, D** and **E** which occupy two tightly linked loci. Alleles of gene D occupy one locus called locus, D while gene C and E alternatively occupy the other locus. The locus is of **prime** importance.

RH" Blood:

Gene D has two alleles, **D** and **d. D** is completely dominated over **d.** Person having genotype **DD** or **Dd** have **Rh factor** on their RBA and are **Rh+. Persons** with genotype **dd** do not have Rh factor and are **Rh---** Unlike the naturally occurring anti-A and anti-B antibodies of ABO-system **anti-Rh antibody,** production requires a stimulus by the human **Rh antigen itself**. (An Rh" person does not produce anti-Rh antibodies unless he is exposed to Rh antigen).

An **Rh^+ donor** is totally incompatible of **Rh^- recipient.** If an **Rh^- person** receives Rh antigen through wrong **Rh+ blood** transfusion, he will begin to produce anti-Rh antibodies against Rh antigens. Rh^- blood, clear of any anti-Rh antibody from a donor who has never been exposed to Rh antigen can b transfused to **RH^+ recipient.**

ERYTHROBLASTOSIS FOETALIS:

(Maternal foetal Rh incompatibility)

Maternal foetal Rh incompatibility results when an **Rh^- woman** married to an **Rh^+ man** conceives a child who is **Rh^+.** If the man's genotype is **DD,** all of their offspring **(Dd)** will be **Rh^+.** If the man's genotype is **Dd**, half of their offspring with **Dd** genotype will be **Rh^+**.

Effect:

If **RBC** of **Rh^+** foetal cross the placental barrier and enter into **Rh^-** mothers blood stream, the mother's immune system reacts to **foetal Rh antigen** stimulus by producing a large number of **anti-Rh antibodies.** When mother's ati-Rh antibodies seep through placenta into blood circulation of foetus, they start **hemolysis** (break down) bursting of RBC of foetus. As this destruction continues, the foetus becomes **anaemic**. The anaemic foetus starts to release **immature erythroblasts** into his blood stream. That is why this hemolytic disease of the new born is called ***erythroblastosis foetalis.***

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Chronic Conditions:

This anemia may lead to **abortion** or still birth. Even if the pregnency continues, the **liver** and **spleen** of foetus **swell** as they rapidly produce RBC. The breakdown product of RBC called **bilirubin** also accumulates in the foetus. Bilirubin damages his **brain cells** and turns his skin and whites of the eyes yellow. This condition is **jaundice**. So the baby if born alive, suffer from severe **hemolytic anemia** and **jaundice.** Such baby's blood should be immediately replaced by Rh blood, free of **anti Rh antibodies.**

Risks of this Condition:

The **first Rh incompatible pregnancy** may not face much problems if very few of foetal antigens cross placenta into maternal circulation and the amount of maternal antibody production is not very high. But when placenta detaches at birth, a large number of foetal cells enter mother's blood stream and stimulate production of large amount of anti-Rh antibodies by the mother. These anti Rh antibodies persist in mother's blood for a long time and are **persistent risk** of the next **Rh^+ foetus.** Rh sebsitization of **Rh^- mother** is avoided by a simple therapy. She is given an injection of **Rh antiserum** during early pregnancy and immediately after birth.

Treatment:

The Rh antibodies in the Rh antiserum will destroy Rh+ of the foetus before they stimulate production of **maternal anti-Rh antibodies.** The injected anti serum disappears before the next pregnancy.

Protection against Rh Incompatibility:

Sometimes a mild ABO incompatibility protects the baby against a more severe Rh incompatibility. If 0 number conceives A' or B' baby, any foetal A or B type RBC entering the mother's blood are quickly destroyed by her **anti A** or **anti B antibodies** before she can form anti-Rh antibodies.

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EPISTASIS

Definition:

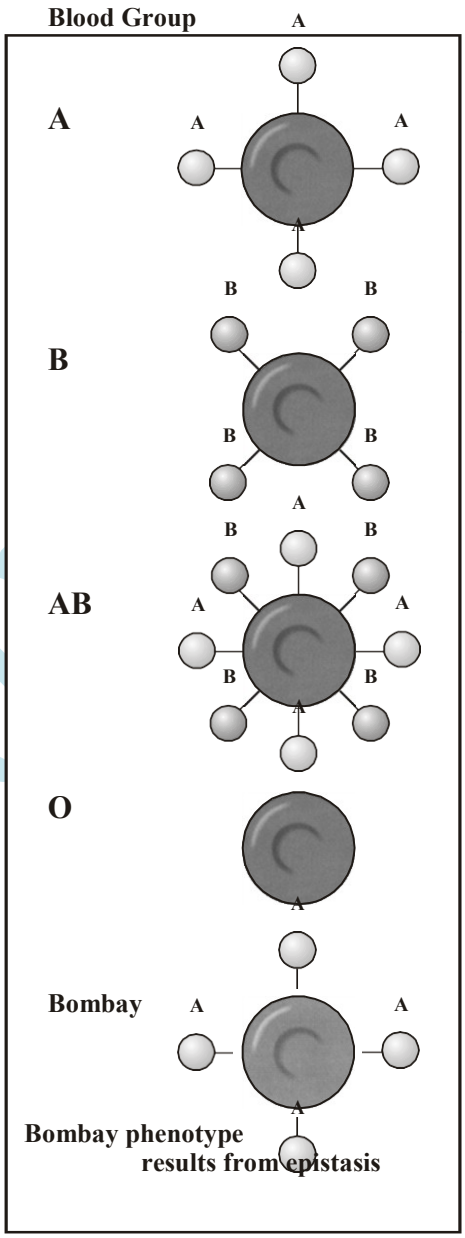
When an effect caused by a gene or gene pair at one locus interferes with or hides the effect caused by another gene or gene pair at another locus such a phenomenon of the gene interaction is called epistasis.

Epistasis and Dominance:

Epistasis must not be confused with dominance. **Dominance** is the relationship between alleles of the same gene occupying the same locus but **epistasis** is the interaction between different gene occupying different **loci.** Following is the example.

Bombay Phenotype:

The expression of ABO type antigen by I^A or I^B gene depends upon the presence of another **gene H.** ABO locus is on chromosome 9, while **H locus** is on **chromosome 19.** H gene changes a precursor substance into **substance H.** It produces an enzyme that inserts a sugar on to a precursor **glycoprotien** on the suface of **RBC**. Only then antigen A or antigen B specified by I^A or I^B gene could attach to this sugar of substance H. The recessive **allele h** cannot insert, sugar molecule to glycoprotein. Therefore, **hh** individuals lack the site of attachment for antigen A or antigen B. Thus A and B antigens cannot adhere to their RBA and **fall away.** Their RBC lack A and B antigens although they do not lack IA and IB genes. They are **phenotypically** like **O,** but are not **genotypically O.** Their phenotype is called Bombay phenotype.



Pleiotropy:

When a single gene affects two or more traits, the phenomenon is called pleiotropy and such genes with multiple phenotypic effect is called pleiotropic.

Example:

1. White eye gene in **Drosophila** also affects the shape of **sperm** storing organs (spermathecae).

2. Genes that affect **growth rate** in humans also influence both **weight** and **height.**

3. In cats the dominant allele W not only makes **fur pure white** but also causes **deafness.** In ww homozygous normal pigmented cats, **melancoytes** produce pigment of fur and also contribute to **hair cells** in inner ear that sense sound. When a cat gets **w allele**, its melonocytes fail to develop properly. Melanocyte failure causes both phenotypes, i.e. white fur and deafness.

CONTINUOUSLY VARYING TRAITS

Genotype interacts with environment to produce **phenotype**.

Phenotypic expression of trait has two aspects.

(i) Quantitative variations (ii) Qualitative variations

(i) Quantitative Variations:

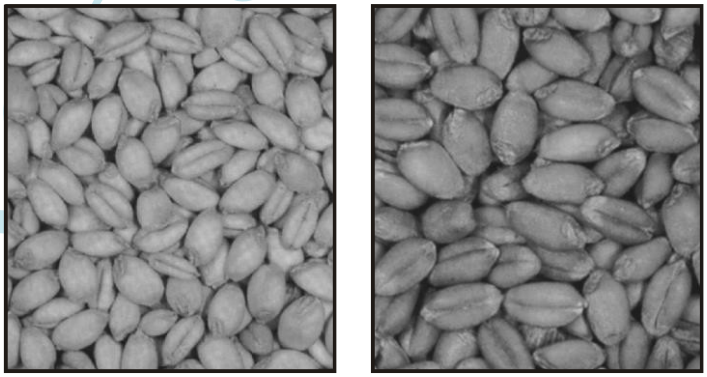
Quantitative variations are small and less striking. Many traits like height, weight, intelligence and skin colour in humans, and grain colour in wheat exhibit continuous quantitative variations over a range of many phenotypes.

(ii) Qualitative Variations:

Qualitative differences are **large** and **more obvious**. Some traits like pea seed shape show discontinuous qualitative variations with two sharply distinct phenotypes, **round** or **wrinkled,** other like **4 O' clock flower colour** can have **three** phenotypes **red, pink** and **white;** still other like **ABO blood groups** system have **four** qualitatively different phenotypes **A,B, AB and 0.**

Polygenic Trait:

Mendel focused on traits that showed only two qualitatively different phenotypes which could be determined by just two alternate alleles of a single gene. **Darwin** observed **small continuous variation** with in individuals of a population. Such a range of phenotypic spectrum of a trait cannot be traced to a single gene with two alleles. Even a few **multiple alleles** of a single gene cannot make such a wide range of phenotypes. A continuously varying trait is encoded by alleles of two or more different gene pairs found at different loci, all influencing the same trait in an **additive way.**



Colour variation in wheat grains is a polygenic trait.

These quantitative traits are called **polygenes traits,** and their genes are **polygenes**. Each polygene has a small positive or negative effect on the character. **Polygenes supplement** each others and sum of positive and negative effects of all individual polygenes produce quantitative phenotypes of a continuously varying trait.

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Different traits of wheat grains:

Wheat grains vary in **colour** from **white** to **dark red**. This trait shows a continuous spectrum of colour variation. Some grains are white, some are deep red but most grains have **shades** in between faint light pink to moderately dark red.

Work of Nilsson Ehle:

Nilsson Ehle studied the genetics of wheat grain colour. When he crossed a **true breeding dark red grain plant** with a true breeding white grain plant, all F1 grain has light red colour, intermediate between two parental shades. It seemed as if it was a case of in complete dominance. But when F1 grains were grown to mature plants and crossed with each other, F2 grains has exactly seven shades of colour in the ratio of I dark red: 6 moderately dark red: 15 red: 20 height red: 15 pink: 6 light pink: I white.

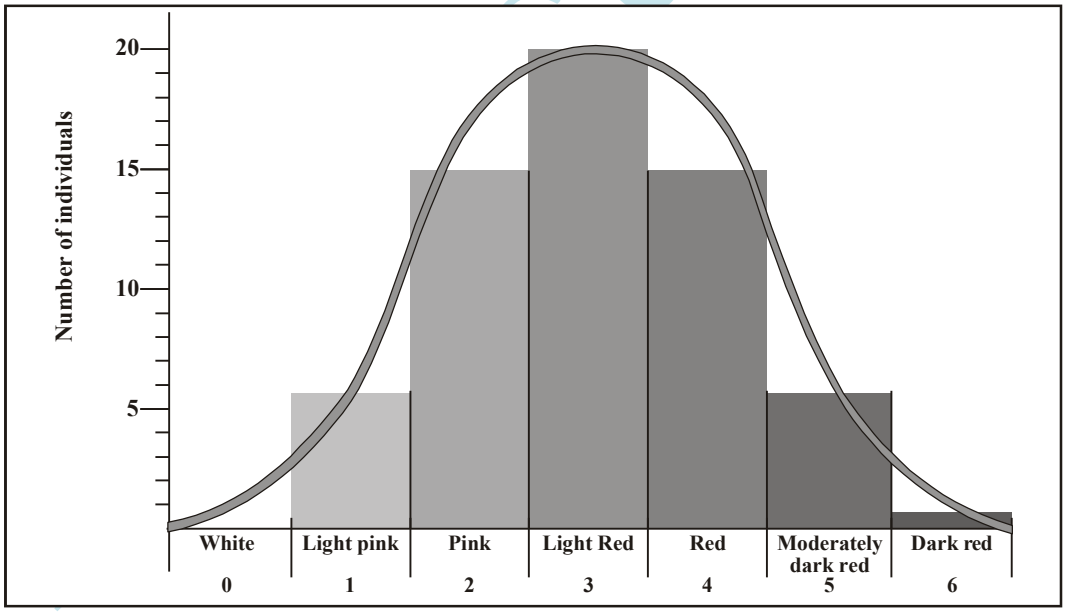
Genetic Basis of Wheat Grain Colour:

Three different gene pairs, i.e. **Aa, Bb, Cc** at three different loci contribute to the wheat grain colour. Each individual would contain **six alleles** for the trait. Alleles **A, B** and **C codes** for an equal amount (dose) of **red pigment,** which is a positive effect. But none of **a, b** and **c** encode red pigment, which is no (zero) dose negative effect.

(a) If all the **six alleles code** for red pigment, **(ABBCC),** the grain is **dark red.**

(b) When **none** of the six alleles encode red pigment **(aabbcc)** the grain is **white.**

(c) When grain has one allele for red pigment (**A**abbcc or aa**B**bcc or aabb**C**c) its colour is **light pink.**



(d) If it has two alleles for the pigment (**A**a**B**bcc or aa**B**b**C**c or **A**abb**C**c) it is **pink.**

(e) If it has **three** pigment alleles (**A**a**B**bCc or **AAB**bcc or **A**abb**CC**) it will be **light red.**

(f) Similarly **four** allele doses (**AABB**cc or aa**BBC**c or **A**abb**CC**) will make **red.**

(g) **Five** alleles colour doses (**AABBC**c or **AAB**c**CC** or (**A**a**BBCC**) produce **moderately dark red** grain.

Thus the colour phenotype of the grain is sum of the individual effect of all the six alleles.

Environmental Factors:

Environemental factors like **light, water** and **nutrients** also influence the amount of grain coluor. Environmental variation make the distribution of phenotypes more smooth and continuous.

Human Skin Colour:

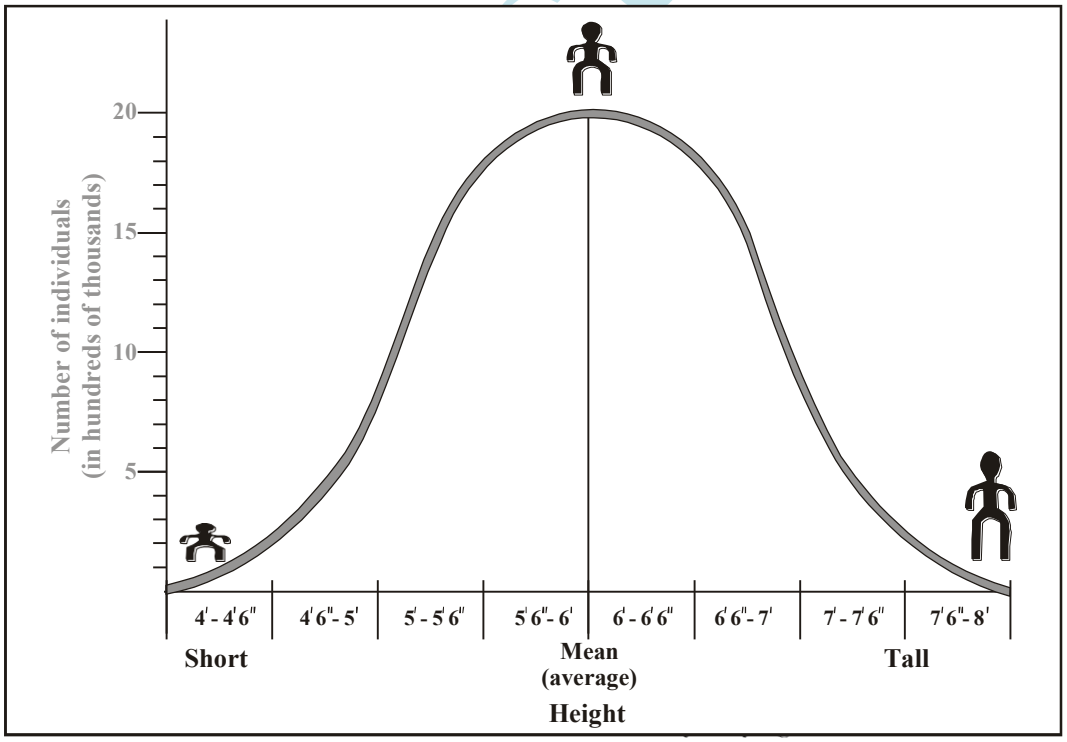
Human skin colour is also **quantitative** trait which is controlled by **three to six gene pairs.** The greater the number of pigment specifying genes, the darker the skin. A child can have darker or lighter skin than his parents.

Human Height:

Human height is a more complex **polygenic trait**. The perfectly continuous variation in range of human heights produces a smooth **bell shaped curve**. A few people are very tall or very short, but most individuals fall in the **average** or **mean** value.

Genetic Basis:

This trait is controlled by many pairs of genes at **different loci.** Even **multiple alleles** may be possible at each locus. More the number of alleles for shortness, the shorter the height will be. Similarly greater the number of alleles for tallness, the taller the height will be.



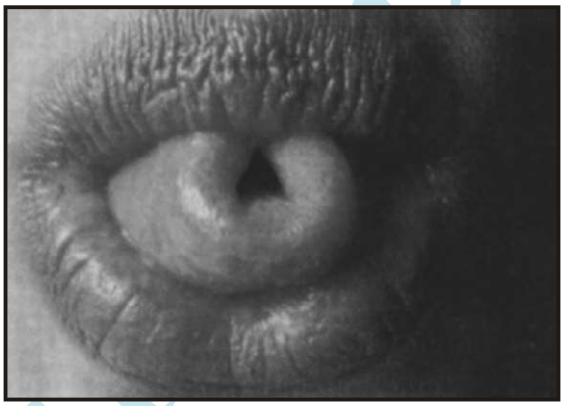
Environmental Factors:

Environment also has a strong **influence** and **skin colour** in humans.

- Constant exposure to **sun darkens skin.**
- **Poor nutrition** prevents achieving genetically determined height.
- **Healthy** and **encouraging** social environment promotes **intelligence.**

Frequency Histogram:

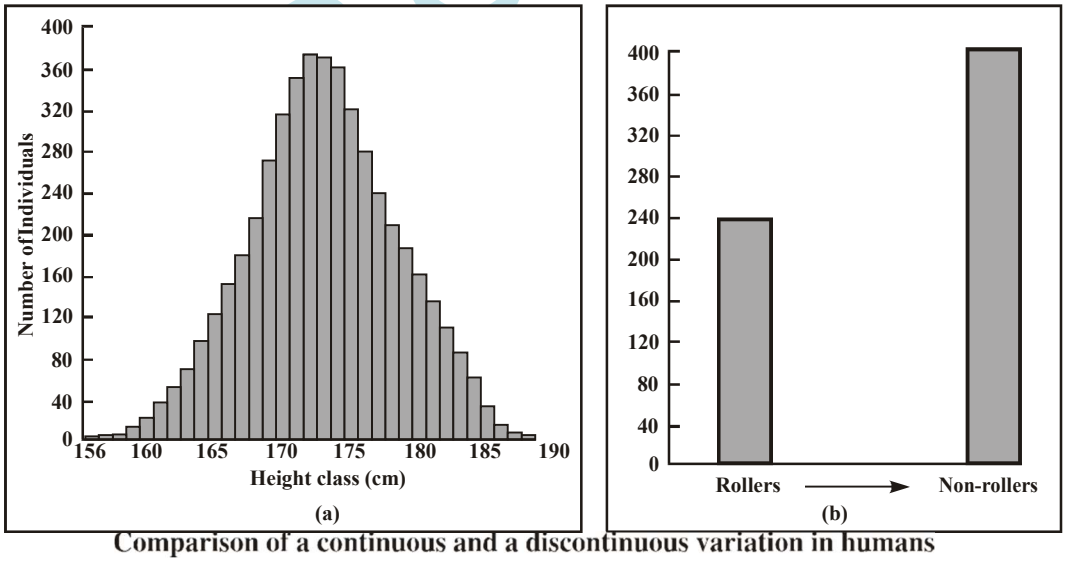
Frequency histograms illustrate **variations**. A frequency histogram is a simple **graph.** The horizontal or **X axis** indicates the **range** to different **phenotypes** of a trait within a population. The vertical or **Y axis** indicates the number of individuals or their **percentage** in a population.



Roller

Tongue rolling:

Some – people can **roll** their **tongue** into a distinct **U shape** when they extend it our of their mouth. They are called **roller.** This ability is due to a single **dominant gene.** It is a discontinuous variation inherited in simple Mendelian fashion. Its frequency diagram from asymmetric distribution curve, with much grater frequency of phenotypes at one end than at the other.



Comparison of a continuous and a discontinuous variation in humans

Frequency Diagram for Human Height:

Human height is a **continuously varying trait.** If we plot a frequency diagram of heights of humans in a large population, so many phenotypes are found with **categories blending** into one another. It forms as **smooth bell shaped** normal distribution **curve.**

GENE LINKAGE

Definition:

The phenomenon of staying together of all the genes of a chromosome is called linkage.

Explanation:

Every organisms possesses numerous characters controlled by thousands of genes, but the number of chromosomes is limited. There for, each chromosome must carry **many genes** on it. All the gene located on the same chromosome are **linked** to each other. **This phenomenon** of staying together of all the genes of a chromosome is called **linkage.**

- Gene linkage is a **physical relationship** between **genes.**
- A chromosome carries its linked genes **en bloc** in the form of a **linkage group.**
- The number of linkage group **corresponds** to the number of **homologous pairs of chromosomes.**
- Linked genes do not obey Mendel's law of independent assortment because these cannot assort independently during meiosis.
- Gene linkages also minimize the chances of genetic recombination and **variations** among **offspring.**

Example:

Man has **23 linkage group.** Genes for **colour blindness, hemophilia, gout** etc. form one linkage group on human **X-chromosome.** Similarly, gene for **sickle cell anaaemia, leukemia** and **albinism** make another linkage group on human **chromosome 11.**

CROSSING OVER

Definition:

Crossing over is an exchange of segments between non-sister chromatids of homologous chromosomes during meiosis.

Process:

The **homologous** chromosomes **pairs up lenthwise,** point to point and locus to locus.

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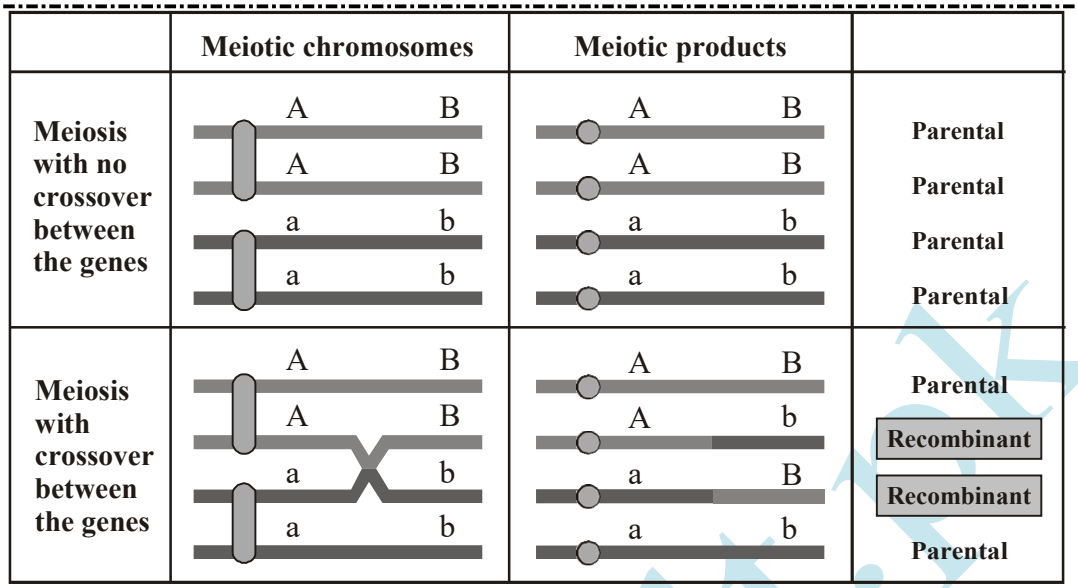

CROSSING OVER OR RECOMBINES GENES

Suppose one homologue carries gene 'A' and 'B' the other homologue has 'a' with 'b' chiasmata are formed at many places between non-sister chromatids f homologous chromosomes. Crossing over occurs at 4 strand stage between non-sister chromatids. It may tae place at more than one place along a chromosome. Exchange of means exchange of DNA, i.e. genes or alleles. As alleles of non-sister chromatids are different, an exchange between their segments results in recombination of genes. Allele 'b' crosses over to chromosomes separate by opening up chiasmata.

The sister chromatids also separate from each other and each becomes an independent chromosome to move singly in each of the four haploid gametes. Four types of gametes are formed two with parental combinations of linked genes i.e., AB and ab, and two with recombination of genes i.e. Ab and aB, (from recombinant gametes). If crossing over does not occur, only the two parental types of gametes are formed. parental types of gametes produce parental types of offspring while recombination gametes produce recombinant types of offspring.

Recombirant Frequency: (Cross over Frequency)

It is the proportion of recombination types between two gene pairs as compared to the sum of all combination.

$$\text{Recombination Frequency} = \frac{\text{Recombinant types}}{\text{Sum of all combinations}} \times 100$$

The recombination frequencies between two linked genes can be calculated by back crossing the heterozygote to a homozygous double recessive recombination frequency is directly proportional to the distance between the linked gene loci. Genes can

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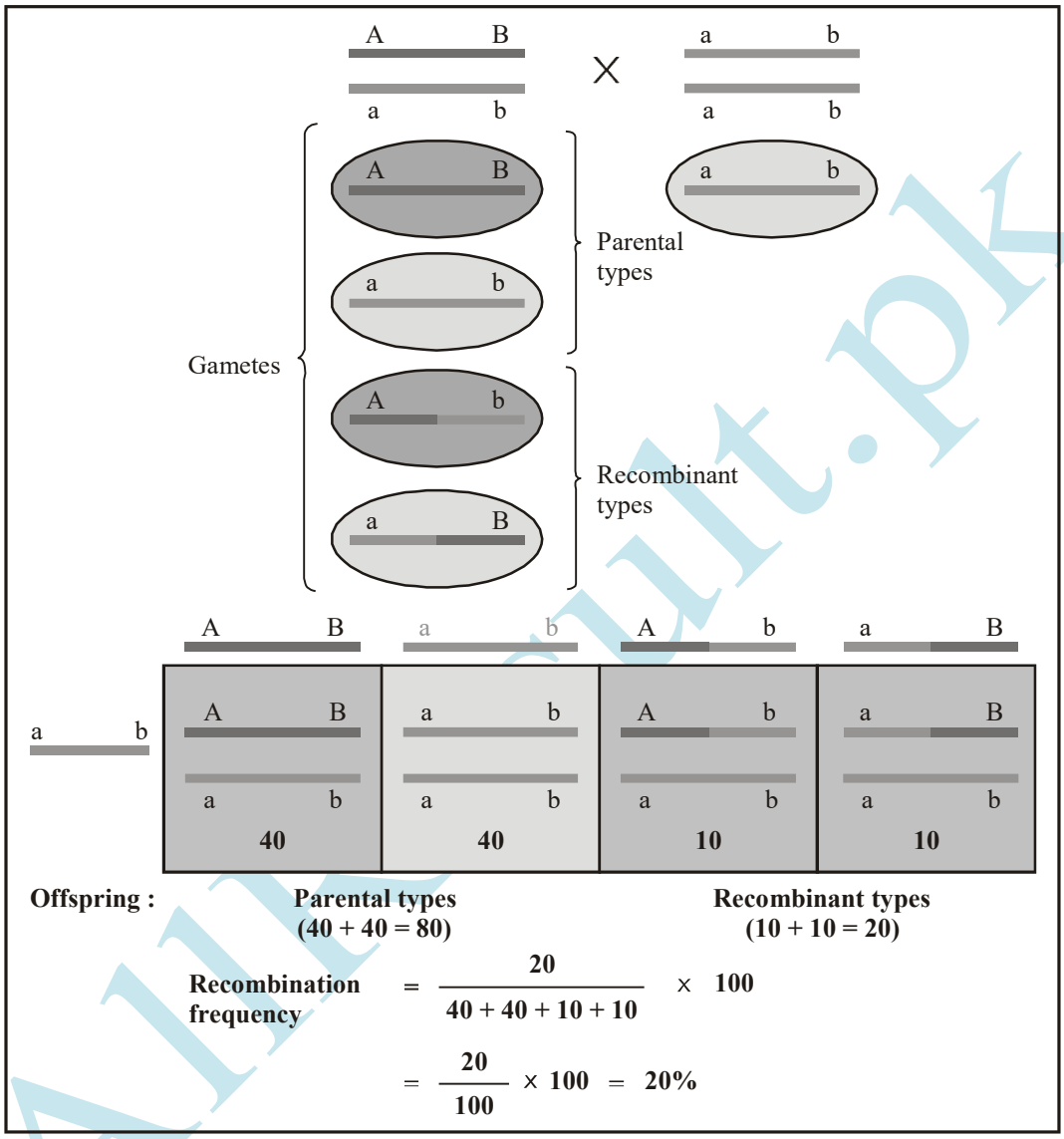
be mapped on a chromosome on the basis of their recombination frequencies. If 1% of recombination frequency is equal to I unit map distance, the two linked genes A and B with a 20% recombination frequency must be 20 units apart.



Importance of Crossing Over:

- Crossing over produce **genetic variation** among offspring.
- Genetic variations lead to tremendous **variations in their traits.**
- Variations provide raw material for evolution by letting them adapt successfully to the changing environment.

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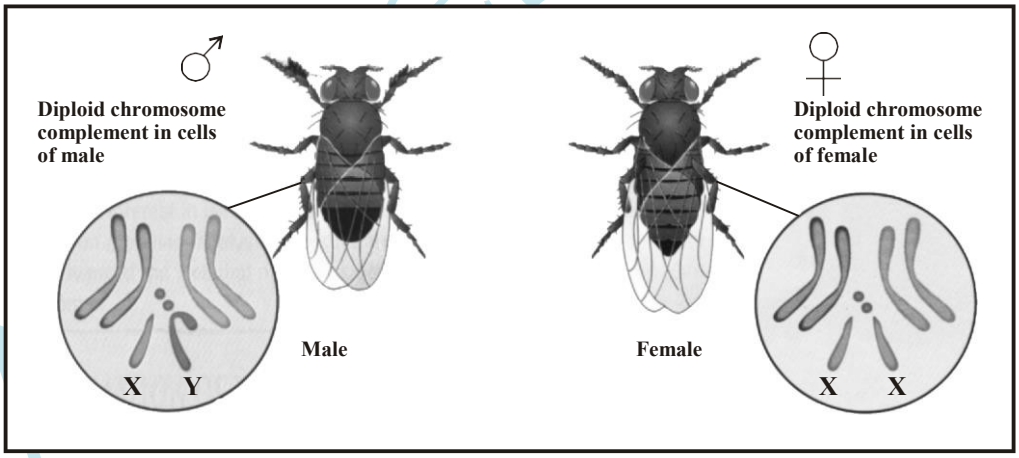
SEX DETERMINATION

Sex Chromosomes:

The chromosomes which determine the sex of the organism are called sex chromosomes. Usually the X and Y determine the sex so they ar called sex chromosomes.

(a) Sex Chromosomes of Drosophila:

Drosophila melanogaster is a **fruit fly** which usually sits on fruits. T.H Moran studied its chromosomal structure. Drosophila has total **four pair of chromosomes.** He found that three pairs of chromosomes were same in male and female **Drosophila** and these are called **autosomes**. The fourth homologous pair of chromosomes was different in both male and female. The female has two similar **X chromosomes** in fourth pair which were **rod shaped**, while **male** has one rod shaped X chromosomes and other morphologically different, J-shaped **Y chromosomes.** These are **sex** chromosomes of ***Drosophila*** as they have genes for determination of **sex**. Chromosomes of the other three pairs are **autosomes.**



Chromosome of Male and Female Drosophila melanogaster

Autosomes:

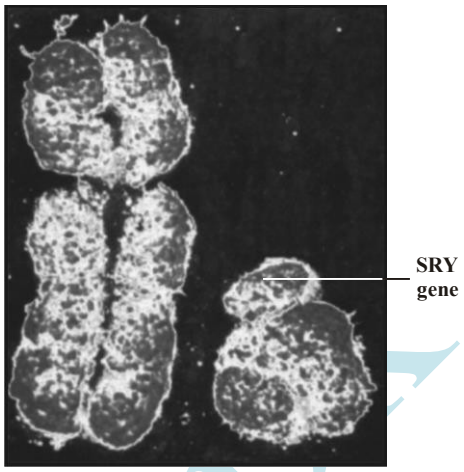
All chromosmes other than sex chromosomes are called autosomes.

Autosomes of not carry any sex determining gene.

(B) Sex Chromosomes in Humans:

In humans there are total **23 pairs** or **46 chromosomes**. Of them **22** are **same** called autosomes while **23rd** pair is sex **chromosome. 23rd** sex chromosome pair is different in males and females. Females are designated as **XX** as it has both similar X chromosome, but in male it is designated **XY** as it has **one X** and other shorter. **Y chromosomes.** The 23rd pair in man is **heteromorepic.**

SRY gene:



Sex chromosomes of man

In **1991 SRY (sex determining regions of T)** gene was discovered on Y chromosomes. It is located on tip of **short arm of Y chromosome.** It **switches on** the male characteristics in **male.**

(c) Sex Chromosomes of Grasshopper:

The sex chromosomes of grasshoppers are entirely **different** from that of other organisms. It has one of the **chromosomes missing in male** which indicates the sex. The total number of chromosomes in **female** is **24** in which there are **11 pairs** of **autosomes** and **one pair of X** chromosomes which designate **female XX.** In male grasshopper there are **total 23** chromosomes in which **11 pairs are autosomes** and there is only **one X chromosomes** which indicates that other chromosome is entirely missing. This **male** is designated as **XO.**

Some species have compound sex chromosomes. They maintain many X or Y or both XY chromosome of more than one kind that act together as a single sex – determining group. That is why the difference in number of chromosomes between male and female is very large. In the round worm Ascaris incurva, the female has 42 chromosomes in the form of 8 pairs of compound X along with 13 pairs of autosomes (16 + 26). Its male has 35 chromosomes comprising 8X plus one Y along with 13 pairs of autosome (8 + 1 + 26).

PATTERNS OF SEX DETERMINATION

Three patterns more common in sex determination.

(1) X 0-XX type
(2) XY-XX type
(3) ZZ-ZW type

1. XO-XX Type:

This pattern of sex determination is found is grasshopper and Protenor bug.

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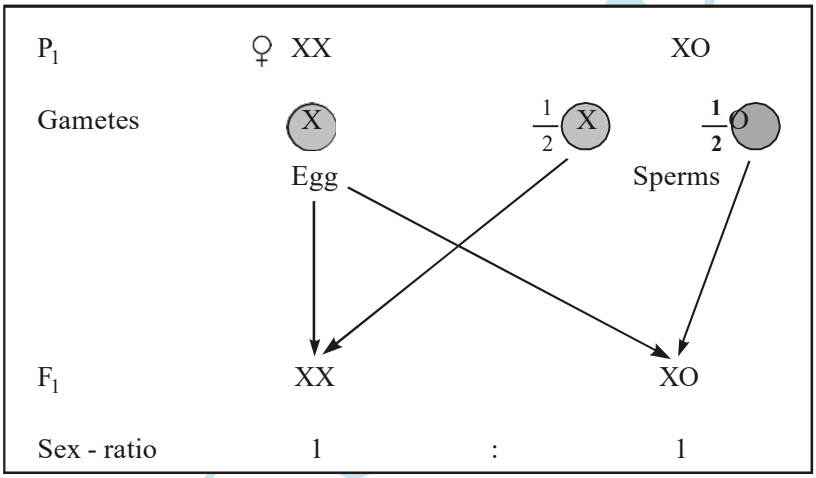
Male Grasshopper:

Male is **XO** because it has only **one X chromosome.** The other sex chromosome is missing entirely. Male is **heterogametic** because it forms two types of sperms; half the sperms have X chromosome while the other **half are without** any sex chromosome. A gamete without any sex chromosome is called **nullo gamete.**

Female Grasshpper:

Female is XX, because it has two X chromosomes. It is homogametic, as it forms only onw type of eggs. Every egg carries a male X chromosomes. Sex of the offspring depends on the kind of sperm that fertilizes the egg. If an X-carrying sperm fertilizes the egg, an XX female offspring is produced. If the nullo sperm fertilizes the egg, an XO male offspring is produced. Sex ratio between male and female offspring is 1:1.



Sex determination in grasshopper and Protenor bug

2. XY-XX TYPE:

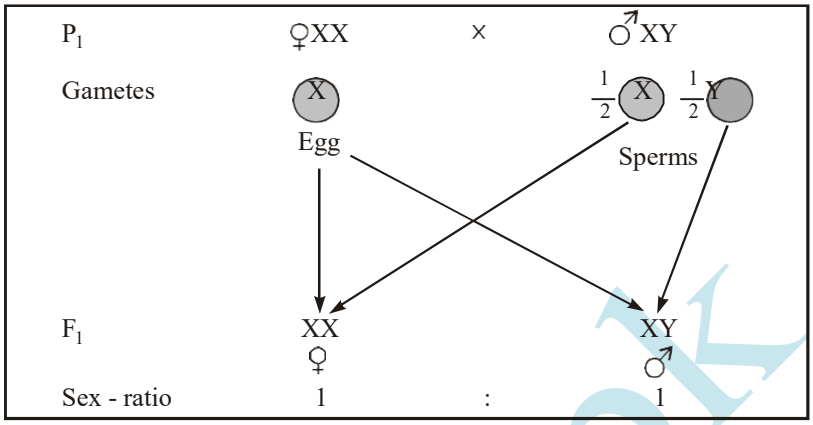
This pattern of sex determination is found in **Drosophila, man** and many other organisms. Male is **XY** and female is **XX.**

Male Chromosomes:

Male being **heterogametic** produces two types of sex-determining **sperms.** Half the sperms carry **X – chromosomes** and the other half carry **Y – chromosome.** Chances for both types of sperms are equal.

Female Chromosomes:

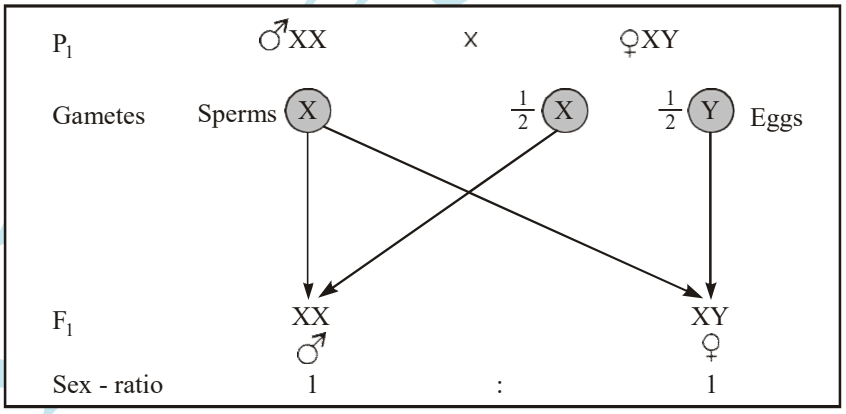
Female being **homogametic** produces only **one type of eggs,** each with an **X chromosome**. Sex of the offspring is determined by the type of **sperm.** If an X – carrying sperm fertilizes the egg, the zygote will be **XY,** and a male offspring will be produced. The sex-ratio between male and female offspring is



Sex determination in man and *Drosophila*

3. ZW – ZZ TYPE:

This type of sex – determination pattern is common in **birds, butterflies** and **moths.** It was discovered by **J. Seiler in 1924** in moth. It is the reverse of **XY – XX** system. Here the female is heterogametic **XY** but the **male is homogametic XX.** Female produces two kinds of eggs X and Y in equal proportions. All sperms are alike, each carrying an X – chromosome. It is the kind of egg that determines the sex of offspring, when an **X – carrying egg** is fertilized by the sperm, a male offspring is produced, but when a **Y – carrying egg** is fertilized by the sperm a **female** offspring is produced. Sex ratio of **1:1.**



Sex determination in birds and butterflies.

COMPARISON OF CHROMOSOMAL DETERMINATION OF SEX BETWEEN DROSOPHILA AND HUMANS

Although both ***Drosophila*** and **humans** follow the same **XY – XX** sex determining **pattern,** yet there is basic technical difference between the two.

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1. Presence of **'SRY'** gene on Y chromosome is essential for **triggering** the development of **maleness in humans.** Absence of Y chromosomes simply leads to the **female development path.**

2. **XO Turner's syndrome** in humans produced through **non – disjunction** is a **sterile female.** But in Drosophila **XO** is a **sterile male.**

3. Similarly **XXY** individual produced through **non disjunctional** gametes in humans is a **sterile male** called **klinefelter's syndrome,** but the same **XXY** set of chromosome in ***Drosophila*** produces a **fertile female.**

4. There is a close genic of different chromosomes. ***Drophila*** has an X **chromosome – autosome** balance system. Its Y chromosomes appears to have very little influence on sex.

5. Here actually the **X chromosme is female determining** and the **autosomes are male determining.**

6. Sex of an individual depends more on the number of X chromosomes relative to the number of **sets of autosomes.** An **X : A ratio** of **1.00 or higher** produces **female** whereas an **X: A ratio** of **0.5 or lower** produces **males.**

Species	XX	XY	X0	XXY
Drosophila	♀	♂	♂	♀
Humans	♀	♂	♀	♂

Comparison of sex determination in man and Drosophila

SEX DETERMINATION IN PLANTS

Plants show a variety of sexual situations.

1. **Separate Sexes:**

Some species like **Ginkgo** are **dioecious** having plants of separate sexes. Male plants produce flowers with only stamens and female plants produce flowers with only carpels.

2. **X – Y System:**

Some dioecious plants have a difference of **sex chromosome** between the sexes.

These have an X – Y system. These plants typically exhibit an **X – chromosome – autosome** balance system for sex determination.

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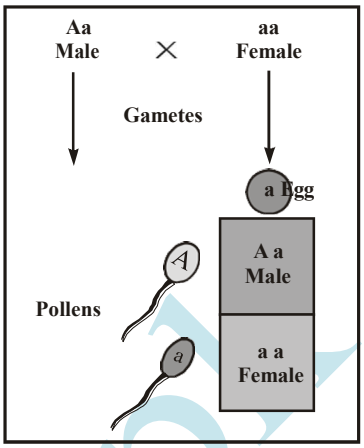
3. Pollens as sex determining:

many other sex determining mechanisms are also seen in dioecious plants. Correns (1907) discovered that pollens of certain plants were sex – determining. All eggs are of one type. Pollens of the two types are produced in equal number. One kinf of pollen after fertilizing the egg produces male plant whereas the other kind of pollen after fertilization produces female plant.



Pellens Determine Sex

4. Genic System:

Many species of eukaryotic micro – organisms like **yeast** do not have **sex chromosome**. These depend on **genic system** for determination of sex. In this system the sexes are specified by simple allelic deference at a small number of gene loci e.g. 'a'; and are the two **mating types** (sexes) of yeast, controlled by **MAT a** and **MAT α alleles** respectively.

SEX LINKAGE IN DROSOPHILA

Drosophila is a very useful organism for genetic studies for many resons:

1. Easily Available:

The tiny fly is often seen hovering over **rotten fruits.** It can be easily collected and cultured on **mashed banana** and other fruits. It does not need large spacious cages. It lives happily in ordinary glass bottle of jams and marmalades. It eats yeast that grows on mashed banana.

2. Sexual Dimorphism:

Male and female ***Drosophila*** shows sexual **dimorphism** i.e. these are morphologically distinct from each other. **Male is smaller** in size with **black rounded abdomen**. **Female is larger** with **pointed abdomen.** Male has **sex combs** on front legs.

3. Short Generation Time:

Drosophila has a generation time of just two weeks. It lays a large number of eggs which hatch out into fertile offspring. Many generations can be raised in a relatively short time.

4. Distinct Contrasting Traits:

Drosophila is perfectly suited for **genetic studies.** It shows fairly large number of **distinct contrasting traits.** Morgan and his colleagues' studied pattern of inheritance of more than **85** traits of *Drosophila.* Its larvae are excellent material for dissection for chromosome study. It has only **eight chromosomes in four homologous pairs** that can be conveniently studied under a microscope. Its **salivary gland cells** have **gaint chromosomes** in their **nuclei**. These gaint chromosome have characteristic banding pattern corresponding to **genes.**

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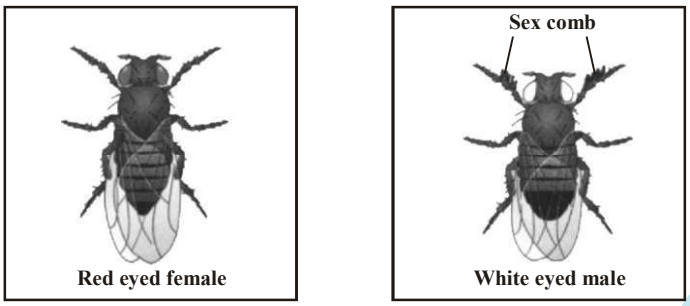
5. Genome is Sequenced:

The **entire genome** of ***Drosophila*** has been successfully sequenced as part of **human genome project.**



Wild type red eyed female and mutant white eyed male *Drosophila*.

Thomus Hunt Morgan (1910) provided experimental evidence in support of **chromosomal theory of heredity** through discovery of sex linkage in fruit-fly ***Drosphila.***

Morgan raised cultures of Drosophila flies to study different traits, such as colour of the eye. Normal fruit flies, the wild type, have **bright red eyes.** One of his coworkers **Calvin Bridges,** observed an unusual **white eye mutant male fly.**

First Step Cross:

Morgan mated this **white eyed male** with a **wild type red eyed female.** All **1237** offspring **(F_1)** of this cross **had red eyes.** Morgan concluded that red eye is a **dominant** trait.

Second Step Cross:

Morgan allows males and females of **F_1 generation** to mate and produce **F2 generation**. He counted **2459 red eyed females, 1,011 red eyed males** and **782 white eyed males** among F_2.

The proportion of **3470 red eyed** to **783 white eyed** flies did not perfectly fit into **Mendelian 3:1 ratio.** The number of recessive phenotype individuals was too small. There was another peculiarity in this result. All the **white eyed flies** were only **males.** There was no **white eyed female** in F_2 generation.

The inheritance of eye colour somehow seemed to be related to the 'sex' of the offspring. **Morgan proposed that**

(i) The gene for eye colour is located on X chromosome.

(ii) The alleles for eye colour are present only on X chromosome. Their is no corresponding allele for this trait on Y chromosome.

Thus even a single recessive allele on X chromosome can express itself in males because Y chromosome is empty for that gene. **Males** are **hemizygous** as they carry just one allele on their only **X chromosome. Female** have two **X chromosome**, each carrying an allele of the trait, Female can be homozygous or heterozygous.

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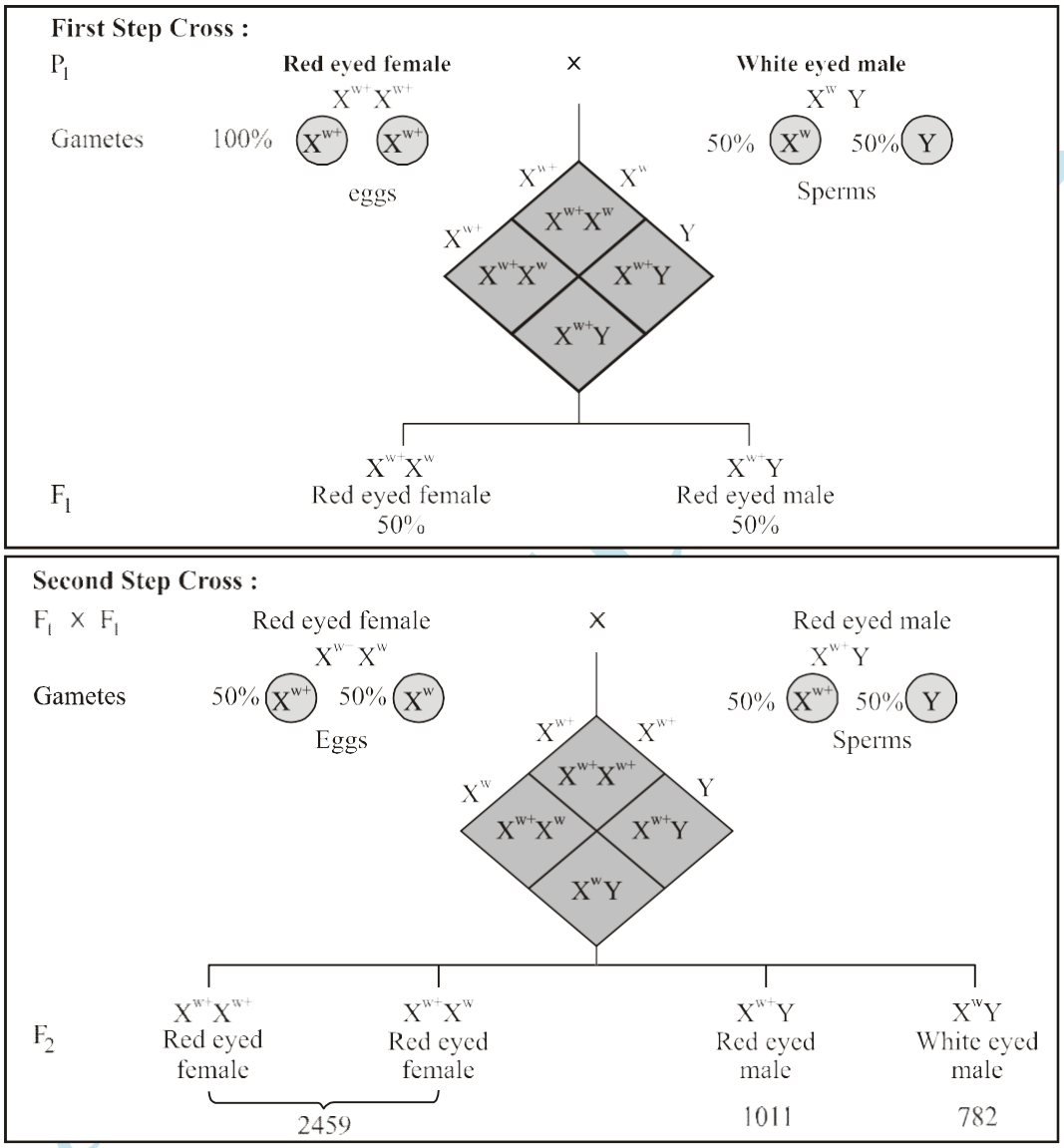
Symbol "w" represents the recessive allele for **white eye,** and **"w^+" designates** its wild type allele for **red eye.** The genotypes of the presents of **P1** cross were: $X^{w+}X^{w+}$ for red eye female, X^wY for the white eye male.

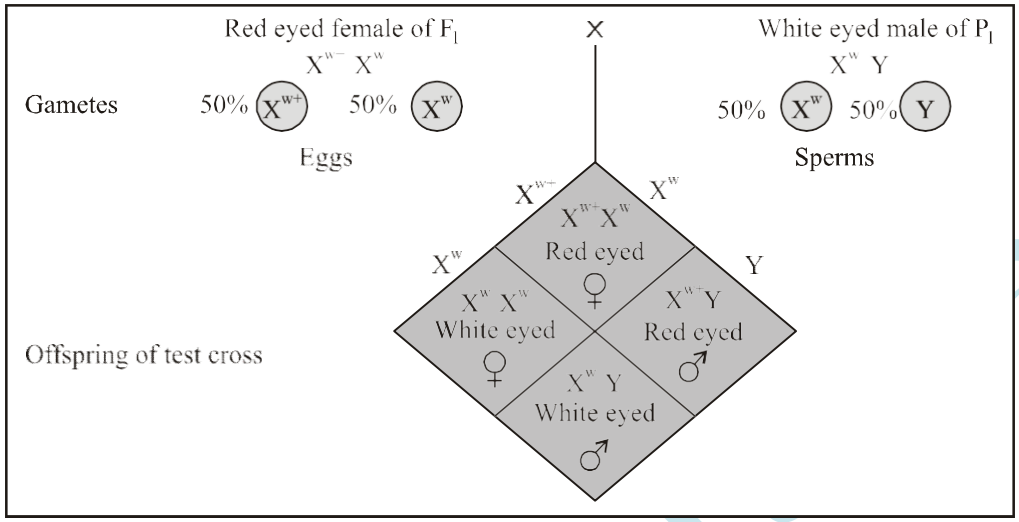
Morgan's hypothesis explained clearly why all the white eyed flies in F_2 generation were only males.



Step 3: Test Cross:

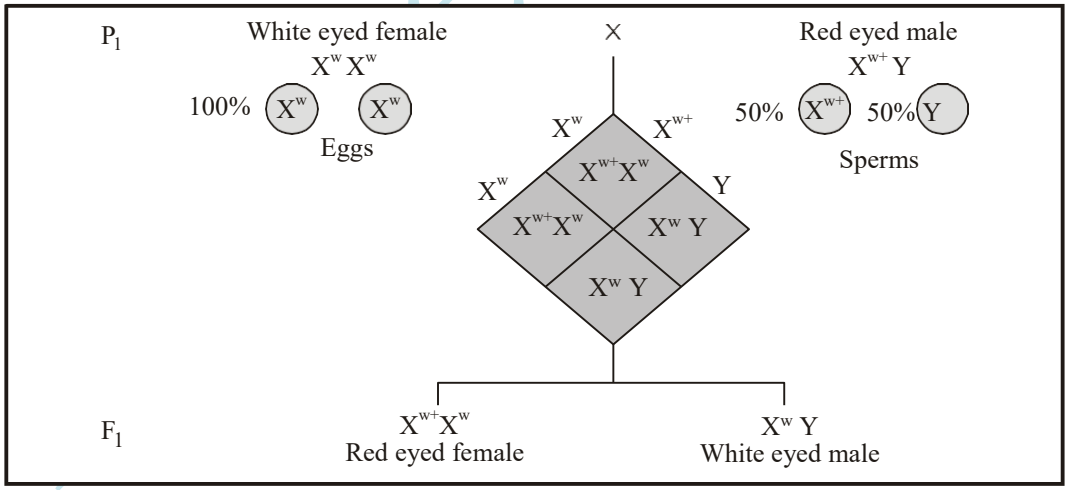
Morgan wanted to test his hypothesis. He crossed the **P_1 white eyed male (X^wY)** with one of its own **daughters**, the **red heterozygous female** form **F_1 generation.** This test cross produced **129 red-eyed females**. **132 red-eyed males**, **88 white-eyed females**

and **86 white-eyed males.** White-eyed flies were **less viable** than red-eyed flies. **Half** the female offspring in fact had red eyes and half had white. Similarly **half** the males had red eyes **half** had white.



Step 4: Reciprocal Cross as a confirmatory Test:

Appearance of white eyed female provided an **opportunity** for a further **confirmatory test.** Morgan mated a **white-eyed female** with **red-eyed male.** All **female** offspring had **red eyes,** and all **male** offspring had **white eyes.** Then these **F_1 red eyed females** and **white eyed males** were **mated** to produce **F_2.** Half of the F_2 females had red eyes, half had white. Similarly half of the F_2 males had red eyes and half had white. This **$F_1 \times F_1$** cross was exactly like **step 3 test cross.**



X Linked Trait:

A trait whose gene is present on X chromosome is called X – linked trait. X – chromosome, having no counterpart on Y chromosome, is called X – liked gene.

Sex – linked inheritance follows a very specific pattern. As a son inherits his **X chromosome** only from his **mother,** and a **daughter** gets an **X chromosome** from **each parent.** An **X – linked trait** passes in a **crisscross** fashion from **maternal grandfather (P_1)** through his **daughter (F_1)** to the **grandson (F2).** It never passes direct from father to son because a son inherits only Y chromosome from father.

Nobel Prize:

Morgan's discovery of **sex – linked inheritance** was a **great contribution** to the understanding of genes and chromosome. In **1933, T.H. Morgan** was **awarded** a Nobel Prize for his contributions to genetics.

Y – Linked Trait:

y chromosome is not completely **inert.** It does carry a **few genes** which have no counterpart on X chromosome. Such genes are called **Y – linked genes** and their traits are called **Y – linked traits e.g. SRY gene on Y chromosome** of man determines **maleness.** Y – linked traits are found only in males. These traits directly pass through Y chromosome from **father to son** only. As females do not normally inherit Y chromosome, such traits can nor pass to them.

X – Y Linked Genes:

Some genes like bobbed gene in *Drosophila* are present on **X and Y** both. These are called **X – and – Y linked genes.** These are also called **pseudoautosomal genes** because their pattern of inheritance is like **autosomal genes.**

SEX – LINKAGE IN HUMANS

Humans have many **X – liked traits** of which some like **haemophila** and **colour blindness** are recessive while other like **hypophosphatemic** or **vitamic D resistant rickets** are **dominant. X – linked dominant** is a trait which is determined by an X – linked recessive gene. Their patterns of inheritance are very different from each other.

Many **X – linked traits** in man are also found **X – linked** in other mammals like **mouse, rabbit, dog, sheep, horse, donkey, cattle, kangaroo** and **chimpanzee.** Was the mamalian X chromosome conserved throughout mammalian evolution?

X – linked recessive inheritance:

Experimental matings are not practically possible in humans. Mode of inheritance of humans trait can be traced through pedigrees.

Genetics of Haemophilia:

Haemophilia is a rare **X – linked recessive** trait. Haemophiliac's **blood fails** to **clot** properly after an injury, because it has either a reduction or malfunction or complete absence of **blood clotting factors.** It is a serious **hereditary disease** because a **haemophiliac** may **bleed to death** even from minor cuts.

Types of Haemophelia:

Haemophilia is of three types: **A, B** and **C.**

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Heamophilia A and B:

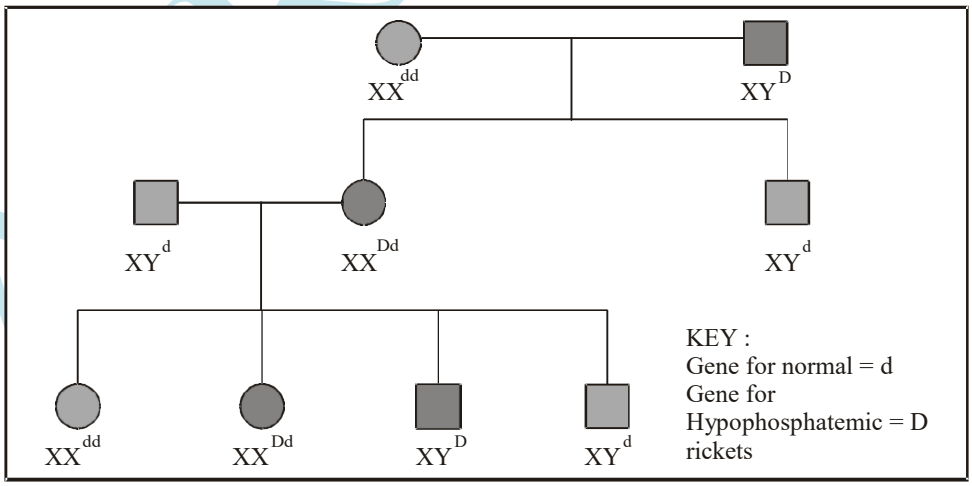
` Haemophilia **A** and **B** are **non – allelic recessive sex – linked, 80& haemophilacs,** suffer from haemphilia A due to abnormality of **factor VIII,** about 20% suffer from haemophilia B due to disturbance in **factor IX.** Being X – linked recessives, haemophilia A and B affect men more than women, Chances for man to be affected by haemophilia A and B are double than woman. A woman can suffer from haemophilia A or B only when she is **homozygous** for the recessive allele, but a man with just on recessive allele will display the trait.

Haemophilia C:

Haemophilia C is an **autosomal recessive trait.** Less than **1%** haemophiliac suffer from **haemphilia C** due to reduction in **factor XI.** Haemophilia **C** affects both the **sexes** equally because it is **autosomal.**

Inheritance of A and B Haemophilia:

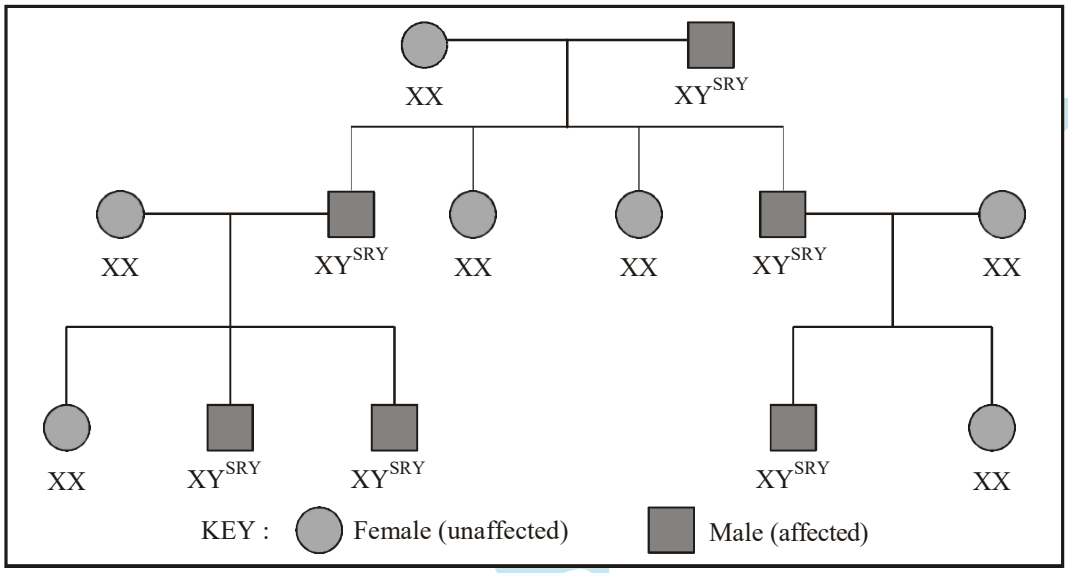
Haemophilia A and B zigzag from **maternal grandfather** through a carrier **daughter** to a **grandson.** It never passes direct from **father to son.** Gene for normal is H, gene for **haemophilia A** is h. in generation I of this pedigree **man (1 – 2)** suffering from **haemophilia A** marries a normal woman **(1 – 1).** He passes haemophilia gene to his **daughter (II – 2)** through his X chromosome. He cannot pass this gene to his son (II – 3) because the son receives only Y chromosome form him. His daughter (II – 2) also receives another X but with normal dominant allele from her mother (1 – 1). The **daughter** looks **phenotypically normal,** but she is **heterozygous** and a **carrier** for the recessive gene. When she marries a normal man **(II – 1)** she passes **her father's trait** to one of her two sons **(III – 4)** who inherits grandfather's X from her. The single recessive **allele** for haemophilia expresses successfully in the **hemizygous** son because his Y chromosome does not carry its **counterpart.** The other son (III – 3) is normal as he inherits grand mother's X with normal gene. One daughter (III – 1) with both normal X is normal, but the other daughter **(III – 2)** is **carrier** like her mother.



Transmission of X-linked dominant trait in humans.

(ii) Y – linked Inheritance:

Pattern of **Y – linked** inheritance is very peculiar. Maleness is a **Y – linked trait.** Y – linked trait passes through **Y – chromosome** from father to son only. Such traits cannot pass to daughter because they do not inherit Y – chromosome. All sons of an affected father are affected by Y – linked trait, **'SRY' gene** on Y chromosome determines **maleness** in **man.** It is male sex **switch** which triggers development process towards maleness after **6 week pregnancy.**



Y-linked inheritance in man.

(iii) Sex Limited Trait:

A sex-limited trait is limited to only one sex due to **anatomical** differences. Such trait affects a structure or function of the body present in only males or only females. These traits may be controlled by **sex-linked** or **autosomal genes.** Genes for **milk yield** in **diary cattle** affect only **cows.** Similarly **beard** growth in humans is limited to **men.** A woman does not grow beard herself but she can pass the genes specifying heavy beard growth to her sons.

(iv) Sex Influenced Trait:

Sex influenced trait occurs in both males and **females** but it is more common in one sex. It is controlled by an allele that is expressed as **dominant** in one sex but **recessive** in the **other.** This difference in expression is due to **hormonal difference** between the sexes. Pattern baldness is a sex influenced trait. many more men than women are bald. It is inherited as an **autosomal dominant** trait in males but as an **autosomal recessive trait in females**. A heterozygous male is bald but a heterozygous female is not. A woman can be bald only when she is homozygous recessive.

Activity:

A man is 45 years old and bald. His wife also has pattern baldness. what is the risk that their son will lose his hair?

DIABETES MELLITUS

Complications of Diabetes:

Diabetes mellitus is a hereditary disease. It is actually a **heterogenous** group of disorders which are characterized by elevated **blood sugar level**. Diabetics are unable to metabolize blood sugar in their body. They pass **glucose** in their urine. Diabetes is the leading cause of **kidney failure**, adult **blindness**, lower **limb amputation** and **heart disease.**

Type of diabetes:

There are two major types of diabetes:

Type I: IDDM (Insulin Dependent Diabetes Mellitus).

Type II: NISSM (Non Insulin Dependent Diabetes Mellitus).

Type I: IDDM

Type I is also called **Juvenile diabetes** because it usually occurs in early age **before 40.** It arises due to deficiency of **pancreatic** hormone **insulin** that normally routes blood glucose to cells for use. Type I is an **auto immune disorder.** The immune system **backfires** and manufactures **auto antibodies** against body's own cells. Sometimes, specific **viral infections** activate auto immune response. **T-cells** (thymus cells) of immune system **attack** pancreas and destroy insulin producing **β—cells**. As a result, pancreas does not produce insulin. Diabetes of type I must receive **exogenous** (from outside source) insulin to survive.

Genetic Basis of Type I:

Progress is being made in understanding the genetic basis of this disease. The insulin gene is located on short arm of **chromosome 11. Polymorphism** and **genetic variations** within this locus is responsible for diabetes type **I susceptibility.** But today, it is no more just a recessive single gene trait, rather it is a **multifactorial** (polygenic with environmental influence) inheritance associated with several alleles.

Type II: NIDDM

Diabetes mellitus **type II** is **non-insulin dependent.** It accounts for **90%** of all diabetic patients. These persons produce some **endogenous** insulin themselves, but their body cells gradually fail to **respond** to **insulin** and cannot take up **glucose from blood**. They develop a sort of **insulin resistance.** It occurs among people over the **age of 40,** and is more common among the **obese. Obesity** increases insulin resistance. As **exercise** reduces obesity it indirectly **increases insulin sensitively** and improves glucose tolerance.

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Genetic Basis of Type II:

There, definitely exists a genetic component in the form of an underlying tendency to develop diabetes under certain **environmental conditions.** About **2% – 5%** of type **II diabetes** get the disease early in life, **before 25 years** of age. It is called **maturity onset diabetes** of the **young (MODY). MODY** can be inherited as an **autosomal dominant trait.** About **50%** of cases of **MODY** are caused by mutation in **glucokinase** gene. **Glucokinase enzyme** usually converts **glucose** to **glucose – 6 – phosphate** in pancreas. MODY can also be caused by mutations in any of the four other genes which encode **transcription factors** involved in pancreatic development and **insulin regulation.** But these four MODY genes do not play any significant role in **adult – onset type II.**

Blood pressure is also an example of **multi-factorial trait.** There is **correlation** between **systolic** and **diastolic** blood pressure of parents and their children. This correlation is partly due to genes common in them. Blood pressure is also influenced by **environmental factors** such as **diet, stress** and **tension.**

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Q.1 Fill in the blanks.

(i) ———— is the basic unit of biological information.

(ii) A sudden change in the structure of a gene is called ————.

(iii) ———— is the chance of an event to occur.

(iv) A cross among monohybrids is a ———— cross.

(v) An individual with a homozygous genotype is called ————.

(vi) Different alleles of a gene that are both expressed in a heterozygote are called ————.

(vii) When a heterozygote exceeds the phenotypic expression of both the homozygotes the phenomenon is called ————.

(viii) When a single gene affects two or more traits, the phenomenon is called ————.

(ix) A gene with multiple phenotypes effect is called ————.

(x) The phenomenon of staying together of all the genes of a chromosome is called ————.

(xi) ———— minimizes the chances of genetic recombination.

(xii) ———— is an exchange of segments between non-sister chromatids of homologous chromosomes during meiosis.

(xiii) All chromosomes other than sex chromosomes are called ———— is the maleness determining gene in man.

(xiv) Type ———— of diabetes mellitus is non-insulin dependent.

(xv) Polygenic inheritance with environmental influence is called ———— inheritance.

ANSWERS

(i) Gene	(ii) Mutation
(iii) Probability	(iv) Monohybrid
(v) Homozygote	(vi) Codominant
(vii) Over-dominance	(viii) Pleiotropy
(ix) Pleiotropic	(x) Linkage
(xi) Crossing over	(xii) Autosomes
(xiii) SRY	(xiv) II
(xv) Multifactorial	

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Q.2 Write whether the statement is true or false. Correct the statement if it is false.

(i) In grasshopper, the male has XY and the female XX types of sex chromosomes.
(ii) Pea is normally a self fertilizing plant.
(iii) Dihybrids are offspring of the parents who differ in one contrasting pair of trait.
(iv) X-linked traits pass direct from father to son.
(v) A person suffering from Blue cone monochromacy can not see blue colour.
(vi) In birds and moths eggs determine sex.
(vii) A hmozygote forms all gametes of the same type.
(viii) The allele for a sex limited trait is dominant in one sex but recessive in the other.
(ix) Pattern baldness is a sex influenced trait.
(x) Carriers of haemophilia show no symptoms of the disease.

ANSWERS

(i)	False	(ii)	True	(iii)	False	(iv)	False
(v)	False	(vi)	True	(vii)	True	(viii)	False
(ix)	True	(x)	True				

Q.3 Encircle the correct answer from the multiple choices.

(i) When a single gene has multiple phenotypic effects, the phenomenon is called:
(a) Codominance (b) Epistasis
(c) Pleiotropy (d) Sex-linkage

(ii) What happens when both alleles of a gene pair independently express in a heterozygote?
(a) Dominance (b) Incomplete dominance
(c) Over dominance (d) Codominance

(iii) A heterozygote offspring quantitatively exceeds the phenotypic expression of both the homozygote parents due to:
(a) Dominance (b) Incomplete dominance
(c) Over dominance (d) Codominance

(iv) How many gene pairs contribute to the wheat grain colour?
(a) One (b) Two
(c) Three (d) Four

(v) Who for the first time found white eye mutant in Drosophila?
(a) Morgan (b) Bridges
(c) Correns (d) De Varies

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(vi) Which of the following traits is transmitted directly from an affected father to only his sons?

(a) Autosomal (b) X-linked
(c) Y-linked (d) X and Y linked

(vii) Which phenomenon reduces the chances of genetic recombination and variations among offspring?

(a) Linkage (b) Crossing over
(c) Independent assortment (d) Dominance

(viii) Which of the following traits is not sex-linked recessive?

(a) Haemophilia (b) Colour blindness
(c) Hypophosphatemic ricket (d) tfm syndrom

(ix) Which of these traits zigzags from maternal grand father through a carrier daughter to a grandson?

(a) Autosomal (b) X-linked
(c) Y-linked (d) X and Y linked

(x) When a haemophlic carrier woman marries a normal man, who among her offspring may be affected:

(a) All her children (b) All her daughters
(c) Half of her daughter (d) Half of her sons

(xi) What is the risk of a colour blind child in a family when mother is colour blind but father is normal?

(a) 100% (b) 75%
(c) 50% (d) 25%

(xii) What is the risk of a colour blind child in a family when father is colour blind but mother is normal?

(a) Zero% (b) 25%
(c) 50% (d) 100%

ANSWERS

(i)	(c)	(ii)	(d)	(iii)	(c)	(iv)	(c)
(v)	(b)	(vi)	(c)	(vii)	(a)	(viii)	(c)
(ix)	(b)	(x)	(d)	(xi)	(c)	(xii)	(a)

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Q.4 Shot Questions.

(i) Differentiate between:

Gene and genome, Phenotype and genotype, Monohybrid and dihybrid, Homozygous and heterozygous, Dominance and epistasis, Autosome and sex chromosome, X – linked trait and Y – linked trait, Allele and multiple allele, Sex limited and sex influenced trait, Incomplete dominance and codominance, Dominant trait and recessive trait, Continuous and discontinuous variatins, Wild type and mutant.

Gene and genome:

Gene is the basic unit of biological information. Hereditary characteristics pass from parents to offspring through genes in their gametes.

The genetic material of an organism is the genome.

Phenotype and genotype:

Phenotype is the appearance of an organism while genotype is the genetic make ip of an organism.

Monohybrid and dihybrid:

A hybrid for a single trait under consideration is said to be monohybrid while a hybrid for two traits under consideration is called dyhybrid.

Homozygous and heterozygous:

When both the alleles of gene pair in an organism are same, the organism is **homozygous** for that gene pair e.g., **'RR'** or **'rr'.**

When the two alleles of a gene pair in an organism are different, the organism is **heterozygous** for that gene pair e.g., **'Rr'**.

Dominance and epistasis:

Dominance is physiological effect of an allele over its partner allele on the same gene locus.

When an effect caused by a gene or gene pair at one locus interferes with or hides the effect caused by another locus, such a phenomenon of gene interaction is called **epistasis.**

Autosome and sex chromosome:

All chromosomes other than sex – chromosomes are called **autosome.** X and Y chromosomes are called **sex – chromosomes** because these have genes for determination of sex.

Allele and multiple allele:

Partners of a gene pair are called alleles. Each **allele** of a gene pair occupies the same gene locus on its respective homologue e.g., RR or Rr etc.

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All the altered alternate forms of a gene, whose number is more than two, are called **multiple alleles** e.g., I^A , I^B , i.

Sex limited and sex influenced trait:

A sex limited trait is limited to only one sex due to anatomical differences. Such trait affects a structure or function of the body present in only males or only females e.g., beard growth in humans is limited to men.

Sex influenced trait occurs in both males and females but it is more common in on sex. It is controlled by an allele that is expressed as dominant in one sex but recessive in the other. This difference in expression is due to hormonal difference between the sexes. Pattern baldness is a sex influenced trait that is more common in men.

Incomplete dominance and codominance:

"When the phenotype of the heterozygote is intermediate between phenotypes of the two homozygotes, it is called incomplete or partial dominance e.g., 4 O-clock pink flower which is the hybrid of white and red 4 O-clock flowers.

Different alleles of a gene that are both expressed in a heterozygous condition are called codominant and the phenomenon is called codominance e., MN blood type.

Dominant and recessive trait:

A trait that appears in a hybrid between two true breeding varieties is called dominant trait while a trait that is suppressed or masked in a hybrid between two true breeding varieties, it is said to be recessive.

Continuous and discontinuous variations:

Some traits show more than two qualitatively different phenotypes, it is called continuous variations e.g., wheat grain colour, human height; skin colour and intelligence.

Wild type and mutant:

An organism with normally existing traits (present in majority of the individuals of the population) is called **wild type** while an organism with a trait developed due to to mutation is said to be **mutant.**

(ii) What is gene pool?

Ans: All the genes / alleles found in a breeding population at a given time are collectively called the **gene pool.** It is the total genetic information encoded in the total genes in a breeding population existing at a given time.

Example: Bean Bag Genetics.

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(iii) Was pea a lucky choice for Mendel? What would have happened if he had studied an eighth character?

Ans: Yea, pea was a lucky choice for Mendel as he studied seven traits and pea plant has seven pairs of chromosomes. The genes of these traits were luckily located on separate chromosomes. The genes of these traits assortment. If he has studied the eighth character, he may have found deviation from independent assortment.

(iv) What is a test cross? Why did Mendel devise this cross?

Ans: Test cross is a mating in which an individual showing a dominant phenotype is crossed with an individual showing its recessive phenotype. This cross finds out the homozygous of heterozygous nature of the genotype. Mendel devised a cross to test the genotype of an individual showing a dominant phenotype.

(v) What would happen if alleles of a pair do not segregate at meiosis? How would if affect the purity of gamete?

Ans: If alleles of a pair do not segregate at meiosis, some gametes have an extra chromosome while others would lack one chromosome. This process is called non-disjunction. This phenomenon disturbs the purity of gametes according to which each gamete should receive only one of the two alleles.

T.c = dominant plant × Recessive Homozygous

(vi) If the alleles do nor assort independently, which type of combination of missing in the progeny.

Ans: The recombinants would be missing in the progeny.

(vii) Why each gamete had equal chance of getting one or the other allele of a pair?

Ans: It is because of meiosis and segregation.

(viii) Does the dominant allele modify the determinative nature of its recessive partner? What short of relationship do they have?

Ans: The dominant allele does not modify the determinative nature of its recessive partner. Dominance is a physiological effect of an allele over its partner allele on the same locus. When one allele is completely dominant over the other, presence of recessive allele is functionally hidden. So the heterozygote has the same phenotype as homozygote.

(ix) Which type of traits can assort independently?

Ans: The trait located on different chromosomes can assort independently.

(x) Why does the blood group phenotype of a person remain constant through out life?

Ans: The blood group phenotype is controlled by genes which will never change or mutate during the life time of a person so blood group phenotype remains constant throughout the life.

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(xi) What is a universal blood donor?

Ans: O blood group individuals are called universal donors. Phenotype O can also be used as donor for small transfusions to A, B and AB recipients because donor's antibodies are quickly absorbed by other tissues or greatly diluted in the recipient's blood stream.

(xii) How can you protect the baby against Rh – incompatibility?

Ans: Sometimes a mild ABO incompatibility protects the baby against a more severe Rh incompatibility. If O mother conceives A^+ or B^+ baby, any foetal A or B type RBC entering the mother's blood are quickly destroyed by her anti – A or anit – B antibodies, before she can form anti – Rh antibodies.

(xiii) Which types of genes do not obey law of independent assortment?

Ans: The genes located on the same chromosome do not obey law of independent.

(xiv) How can linked genes be separated from each other?

Ans: The linked genes can be separated from each other by crossing over.

(xv) What is multifactorial inheritance?

Ans: The inheritance of a trait which is controlled by several genes and is affected by environmental factors as well is called multifactorial (polygenic with environmental influence inheritance.

Example: Blood Pressure.

(xvi) What is MODY?

Ans: See text.

(xvii) Can a child have more intelligence (IQ score) than his parens.

Ans: Yes, a child many have more intelligence (IQ score) than his parents.

Q.5 Extensive Questions

(i) What is incomplete dominance? Explain it with example.

Ans: See text.

(ii) Define Mendel's law of segregation. Explain it with an example.

Ans: See text.

(iii) Define Mendel's law of segregation. Explain it with an example.

Ans: See text.

(iv) Define probability. Derive 9:3:3:1 F2 ratio of independent assortment though product rule.

Ans: See text.

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(v) What is codominance? Explain the phenomenon of codominance with an example.

Ans: See text.

(vi) Define multiple alleles. Describe multiple allelic blood group system of man.

Ans: See text.

(vii) What is Rh – factor? Describe the genetic basis of Rh – blood group system of man.

Ans: See text.

(viii) What is erythroblastosis foetalis? Discuss this adverse effect of Rh – incompatibility? Also suggest a therapy to avoid Rh sensitization of an Rh – mother married to an Rh+ man.

Ans: See text.

(ix) Define epistasis. Explain epistatic gene interaction with an example.

Ans: See text.

(x) What is a pleiotropic gene? Discuss pleiotropy with examples.

Ans: See text.

(xi) What are polygenes? Explain polygenic inheritance.

Ans: See text.

(xii) What is crossing over? Define recombination frequency and explain its significance.

Ans: See text.

(xiii) What are sex – chromosomes? Discuss the chromosomal patterns of sex determination in organisms.

Ans: See text.

(xiv) Compare chromosomal determination of sex between Drosophila and humans.

Ans: See text.

(xv) Define gene pool. Explain the concept of gene pool in a sample of human population.

Ans: See text.

(xvi) What is sex linkage. Explain T. H. Morgan's study of sex – linkage in Drosopila.

Ans: See text.

(xvii) Compare the pattern of inheritance of an X – linked dominant trait with an X – linked recessive trait in humans.

Ans: See text.

(xviii) Explain diabetes mellitus and its genetic basis.

Ans: See text.

(xix) Discuss the genetics of colour blindness or haemophilia.

Ans: See text.

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