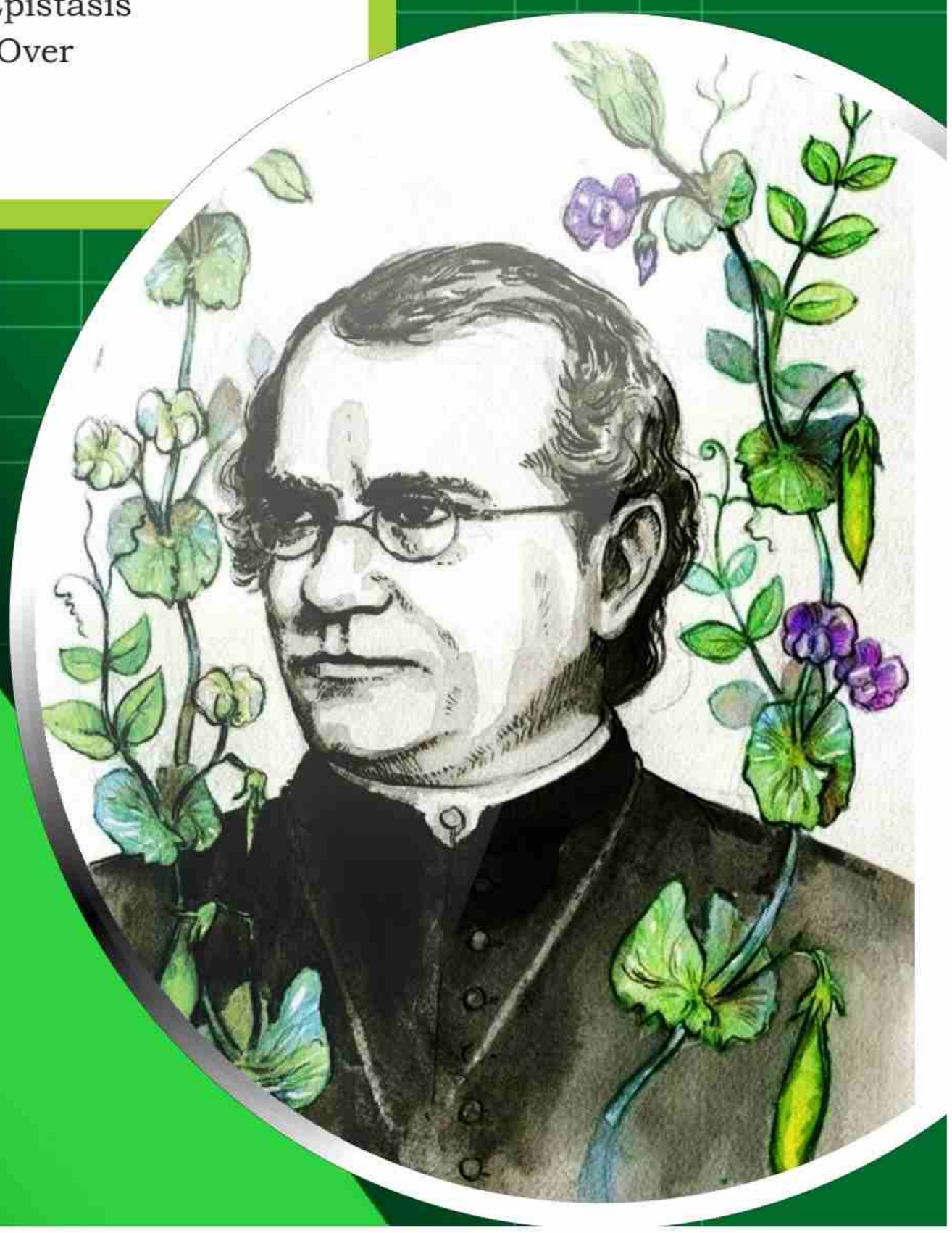
# INHERITANCE

Chapter 23

## Major Concept

**In this Unit you will learn:**

- Laws of Mendel
- Incomplete Dominance, Multiple Allele and Co-dominance
- ABO Blood group system.
- Rh Blood Type System and Erythroblastosis foetalis
- Polygenic Inheritance and Epistasis
- Gene linkage and Crossing Over
- Sex Determination
- Sex Linkage.



We have studied in early classes that the scientific study of inherited characters from parents to offspring, their pattern of inheritance and causes of variation called **Genetics**. In the course of history the simplicity was replaced by complex questions like what factors account for similarities between generation and varieties? What is inherited and what is not inherited? What heredity factors do members of species have in common and in what factors do they differ? How we can control heredity? These questions are addressed by Mendelian, non-Mendelian and modern advancement in genetics.

## 23.1 LAWS OF MENDEL

The earlier work on hybridization experiments on plant were failed because of their methodology and choice of material **Gregor John Mendel** an Austrian Monk succeeded to organize experiments and got logical results from them. The reason of his success lied in his choice of material and method of study. He chose *Pisum sativum*, because it is easy to cultivate and self-pollinated plant. It also contains big sized flower where cross-pollination can artificially takes place. The Pisum plant has number of varieties with sharply defined contrasting characters. As a result of cross-pollination they produce fertile hybrid offspring. He was not pure biologist so he avoided the complexities which troubled the earlier workers.

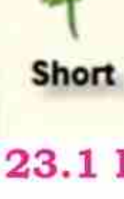
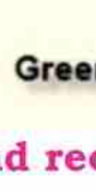
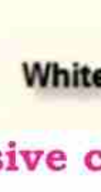
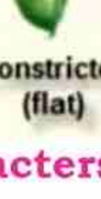
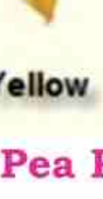
| | Height | Seed Shape | Seed Color | Seed Coat Color | Pod Shape | Pod Color | Flower Position |
|---|---|---|---|---|---|---|---|
| Dominant |  Tall |  Round |  Yellow |  Green |  Inflated (full) |  Green |  Axial |
| Recessive Trait |  Short |  Wrinkled |  Green |  White |  Constricted (flat) |  Yellow |  Terminal |

**Fig. 23.1 Dominant and recessive characters of Pea Plant**

Mendel started his studies by planting seeds with different contrasting characters which he selected for his studies. As a result he got pure breed of these plants. Then he planted the seed of these pure plants, at the time of flowering he crossed plants of contrasting characters to see the pattern of inheritance in the next generation, for example, he crossed smooth seeds producing plant with wrinkled seed producing plant. In this way he crossed plants of seven contracting characters plants as given in table 23.1.

**Table: 23.1 Contrasting characters in pea plant**

| S. No | Cross between contrasting characters | Results of 1st Generation (F1) | Results of 2nd Generation (F2) | Ratio in F2 |
|---|---|---|---|---|
| **1** | Tall x dwarf | All Tall | 787 Tall<br>277 dwarf | 2.84:1 |
| **2** | Round seed x wrinkled seed | All Round seed | 5474 Round<br>1850 wrinkled | 2.96:1 |
| **3** | Yellow cotyledon x green cotyledon | All Yellow cotyledon | 6022 Yellow<br>2001 green | 3.01:1 |
| **4** | Purple flowers x white flowers | All Purple flower | 705 Purple<br>224 white | 3.15:1 |
| **5** | Smooth pods x constricted pods | All Smooth pods | 882 Smooth<br>299 constricted | 2.95:1 |
| **6** | Green pods x yellow pods | All Green pods | 428 Green<br>152 yellow | 2.82:1 |
| **7** | Axial flower x terminal flower | All Axial flowers | 651 Axial<br>207 terminal | 3.14:1 |

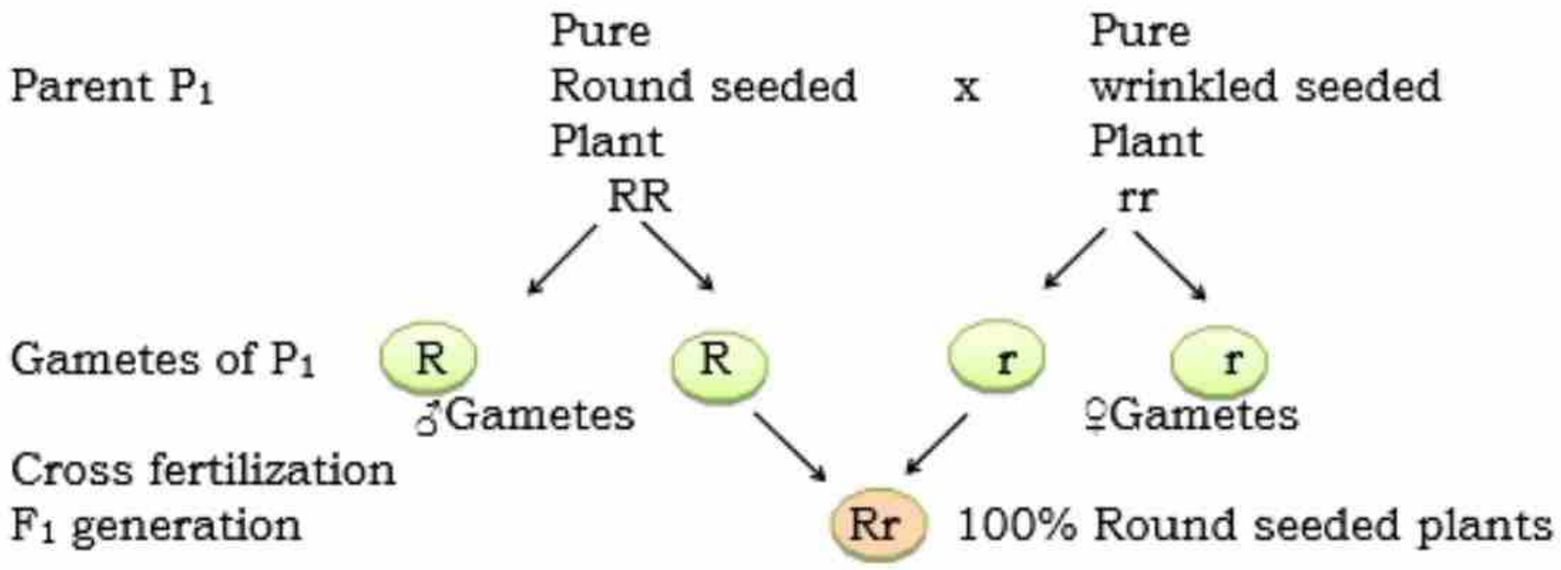
After crossing these plants he observed the result in 1st generation and maintained the record of data for each cross. He came to the conclusion that when pea plants with two contrasting expression of same character were crossed, one of the two expressed completely in the offspring while the other did not expressed at all. From this observation he derived the **law of dominance** which states that in **hybrid** (impure) condition only one character express, completely while other is masked completely. The character which was expressed in impure condition (1st generation, F1) called **Dominant** and its contrasting character, which was not expressed called **recessive**. He found same results in all studied characters of pea plant.

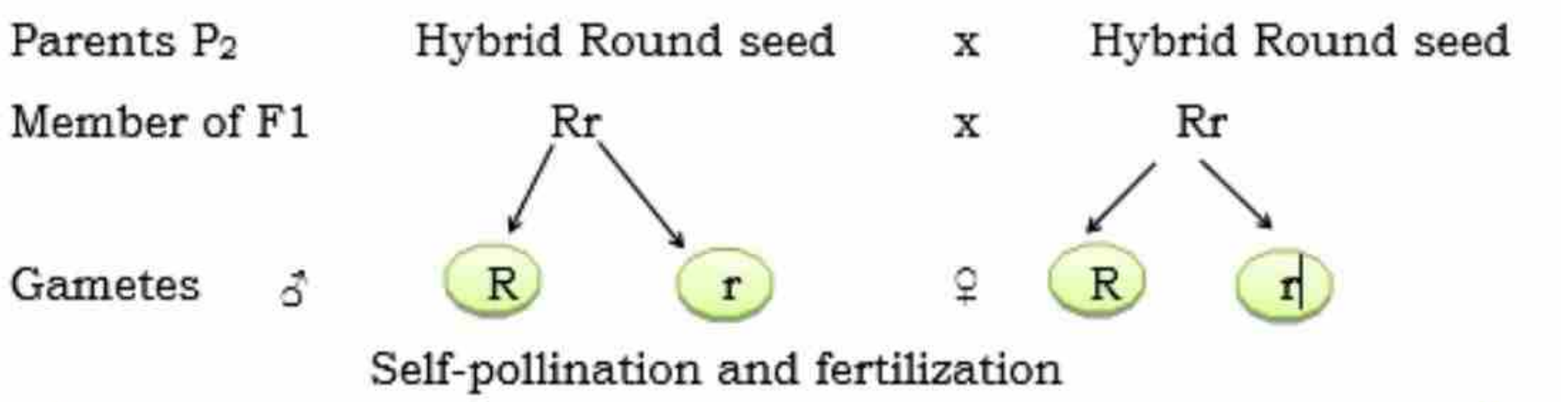
Mendel then proceeded to sow the hybrid seeds of the first generation (F1; Filial generation) and the plants were allowed to self-pollination for getting seeds of second generation (F2). When he sow these seeds he found that in F2 generation the dominant and recessive both appeared in the ration of 3:1. He also discovered that whatever the character pair he studied, the ration of plants with dominant character to those with recessive characters was always close to 3:1 as shown in table 23.1.

Mendel also explained that each organism contains two **factors** for each trait, one is **paternal** and other is **maternal**. They may be similar or different depend on parents. Due to logical mind Mendel gave a generalized theoretical explanation of his results obtained from these experiments in the form of law called **Law of Segregation**.

### 23.1.1 Law of Segregation

The law of segregation deals with the hybridization of only one character (trait) therefore the crosses involved in this experiment called monohybrid crosses. In this way it explains the inheritance of single trait. The law states that factors of each trait segregate cleanly during the formation of gametes so that each gamete contains only one factor of a trait. This law is also called **law of purity of gametes**, because each gamete contains only one factor of a trait.



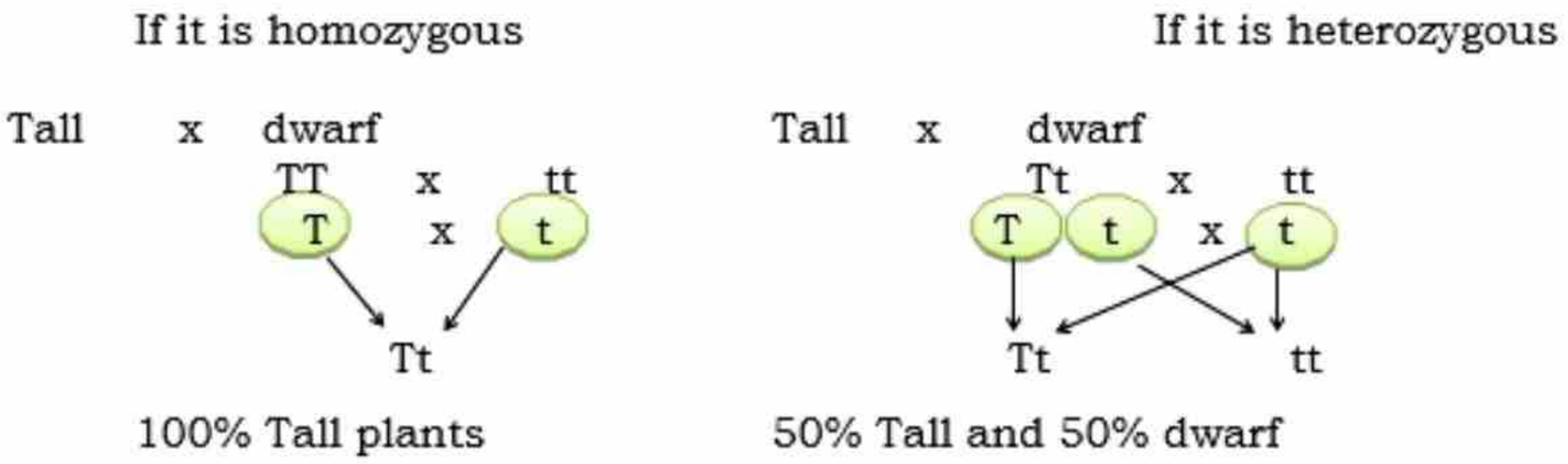




| ♂ / ♀ | **R** | **r** |
|---|---|---|
| **R** | RR<br>Pure Round seed | Rr<br>Hybrid Round seed |
| **r** | Rr<br>Hybrid Round seed | rr<br>pure wrinkled seed |

Now when are using some different terms from Mendalian terms like factors for genes and trait for character, **homozygous** for pure breed of a trait, **heterozygous** for hybrid or impure trait, for physical appearance of a trait we use **phenotype** and the pair of donated factors by parents for a trait called **genotype**. For example, Round seeded homozygous is RR while Round seeded heterozygous plant has genotype Rr.

During studies of single trait inheritance he found that if plants of heterozygous genotype are self-pollinated produce two types of plant, i.e. dominant and recessive, with the phenotypic ratio of 3:1 respectively. Genotypically they have ratio of 1:2:1 i.e. homozygous round seeded: heterozygous round seeded: homozygous wrinkled seeded.

Sometime it makes confusion that the phenotypically dominant individual is either homozygous or heterozygous because both are similar in their appearance. This confusion can be resolved by a cross called **Test cross**. This cross is made between phenotypically dominant individual and homozygous recessive individual to find out the **homozygosity** or **heterozygosity** of dominant parent. When dominant homozygous parent is crossed with recessive, all the offspring will be dominant phenotypically but when cross with heterozygous dominant then half one of the offspring will be dominant and half recessive, as follows




i.e 1:1 ratio.

## Inheritance of Two Traits

Mendel experiment on garden peas were not limited to single traits but sometimes involved two or more traits. After studying inheritance pattern of single trait Mendel studied two trait inheritance patterns to find out the inheritance pattern of different traits of an organism at same time. He crossed pea plant having two traits of contrasting varieties. This cross is called di-hybrid cross and the result obtained as a result of these crosses called **Dihybrid ratio**.

Mendel chose a pure tall pea plant which produces round seed and a pure dwarf pea plant produces wrinkled seed cross together by artificial means. He found all tall plants which produce round seed, from these results he infer perhaps these two assorted traits of a plant are dependent on each other during inheritance because only one of the parental combination is produced in F1 generation. When he left the members of $F_1$ generation for self-pollination and get their offspring he found some new combinations which are different from parental combination i.e. some tall plants can produce wrinkled seed

or some dwarf plants can produce round seed as shown in flowing cross.

Parent ($P_1$) — Tall plant produce Round wrinkled seed homozygous for both TTRR — x — dwarf plant produce seed homozygous for both ttrr

Gametes of $P_1$ — TR, tr

$F_1$ generation — TtRr

All Tall and produce Round seeds

Self-pollination

TtRr x TtRr

Gametes — TR Tr tR tr x TR Tr tR tr

| TR | TR | Tr | tR | tr |
|---|---|---|---|---|
| TR | TTRR Tall & Round seed | TTRr Tall & Round seed | TtRR Tall & Round seed | TtRr Tall & Round seed |
| Tr | TTRr Tall & Round seed | TTrr Tall & wrinkled seed | TtRr Tall & Round seed | Ttrr Tall & wrinkled |
| tR | TtRR Tall & Round seed | TtRr Tall & Round seed | ttRR dwarf & Round seed | ttRr dwarf & Round seed |
| tr | TtRr Tall & Round seed | Ttrr Tall & wrinkled seed | ttRr dwarf & Round seed | ttrr dwarf & wrinkled seed |

This experiment revealed that in di-hybrid crosses some phenotypically new plants i.e. plants with non-parental combinations are also produced. According to above example tall plants do not

produce round seeds like parent they may produce wrinkled seed as well as some plant similarly dwarf plants do not produce wrinkled seeds they may produce round seeds in other dwarf plants. In this way Mendel found some parental and some non-parental combinations in $F_2$ ration as follows

| Tall plants producing: Round seeds (Parental) | Tall plants producing: wrinkled seed (Non-Parental) | Tall plants producing: Round seeds (Non-Parental) | Tall plants producing: wrinkled seed (Parental) |
|---|---|---|---|
| $\frac{9}{16}$ | $\frac{3}{16}$ | $\frac{3}{16}$ | $\frac{1}{16}$ |

Above results led him to the formulation of a law called **Law of Independent assortment** which states that "The factors of assorted traits are independent in their inheritance". OR

The members of one pair of gene segregate independently of the other.

### Extra Reading Material

In an experiment Mendel found Green seed colour (G) is dominant over yellow (g). Find out genotype of following parents whose phenotypes are given F1

| Parents | Green | Yellow |
|---|---|---|
| i) Green x yellow | 82 | 78 |
| ii) Green x Green | 118 | 39 |
| iii) White x yellow | 0 | 50 |
| iv) Green x yellow | 74 | 0 |
| v) Green x Green | 90 | 0 |

**Exception to the Mendel's Law of inheritance.**

Basically Mendel was a Monk, his knowledge of inheritance was based on the experimental results which he observed during cross breeding between different strains of pea plants. In this way he had given basic concept of genes, alleles, dominant and recessive allele. His understanding was limited, up to his observation that each

trait has only two alternating forms i.e. **Allele** or when these two contrasting allele of a trait gather in an organism only one expresses while other does not, which he said **complete dominance**. After Mendel, breeding experiments were carried in different plants and animals where many exceptions were also found. As a result of these cross-breeding experiments, different types of phenotypes and phenotypic ratio were observed which were different from Mendelian ratio. These exceptions were named as non-Mendalian inheritance i.e. incomplete dominance, co-dominance, multiple alleles etc.

## Incomplete Dominance

Carl Correns in 1889 worked on 4 o'clock plants (*Mirabillis jalapa*), During his cross-breeding experiment of pure breed Red flowering plant with white flowering plant he found pink flowering plants. This studies of inheritance showed that many trait have not showed that Mendalian behavior of complete dominance because the 4 o'clock plant in F1 generation had not produced any of the parent phenotype i.e Red or white. Furthermore, when Correns self-fertilized F1 pink flowered plant, the F2 generation showed all three phenotypes of flowers in the ratio of 1(Red) : 2(Pink) : 1(white), the Red and white were homozygous while the pink were heterozygous genotypically.

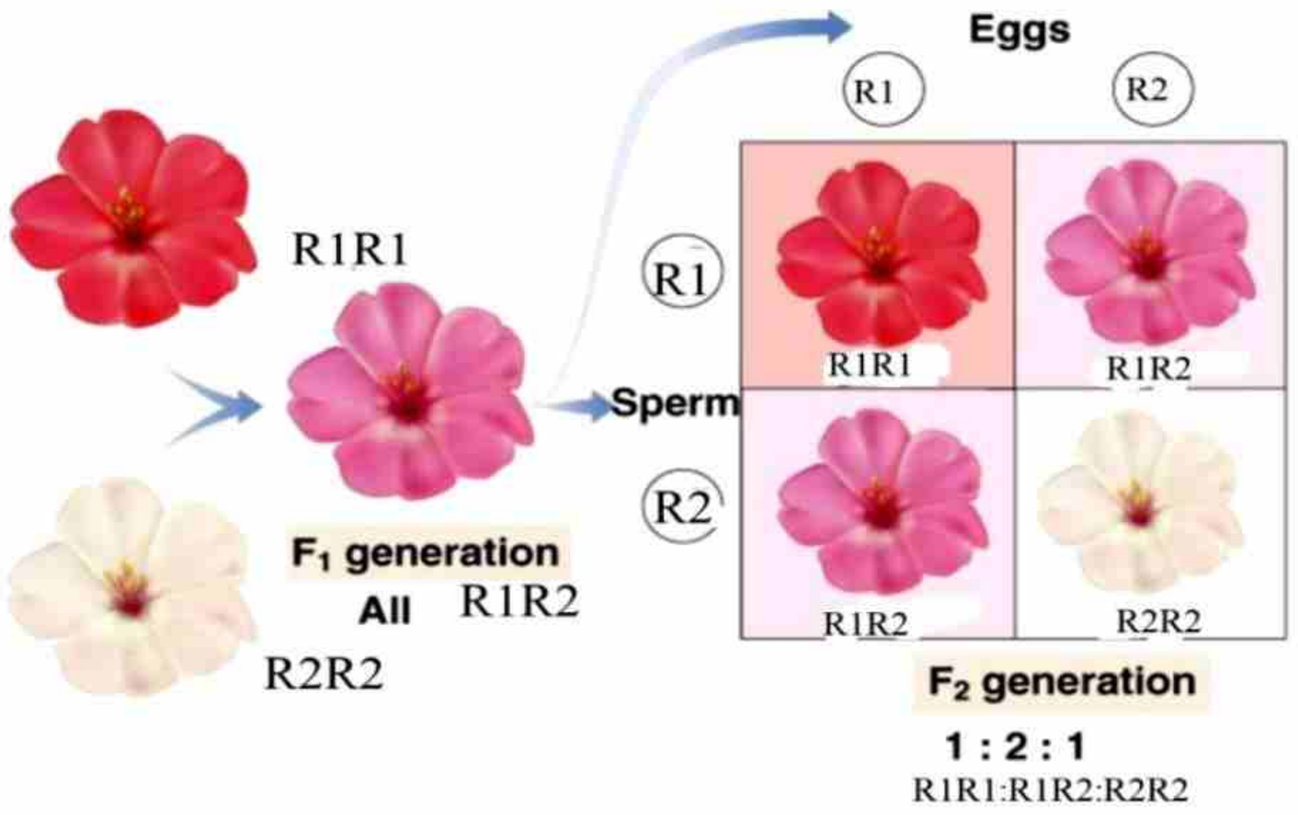



**Fig. 23.2 Incomplete dominance**

It showed that when contrasting allele exist in an individual none of them masked the expression of other, both of them tried to express but these expressions get blended to produce a new phenotype. So he stated that in heterozygous condition both the contrasting allele express and expression get blended to form a new phenotype is called incomplete or partial dominance.

The capital and small letters are used for complete dominant and complete recessive alleles respectively but in the case of incomplete dominance none of the allele is completely dominant over the other so alleles are represented by same letters with sub-script number to distinguish them from each other e.g. for Red flowers ($R_1$) and White flowers ($R_2$).

## Co-Dominance

Another type of deviation from Mendalian complete dominance is co-dominance, where heterozygous individuals where both contrasting alleles of same locus express independently and form their respective products, these products show their expression clearly and independently without any blending.

It may be described as phenomenon of inheritance in which both contrasting allele of a trait are dominant and express themselves in heterozygous individual neither masking nor blending the effect of one another.

In a cross between a true breeding short horn red cattle and a true breeding short horn white cattle, the offspring have roan colour, a close examination of skin of roan coloured animal shows that the animal does not possess an intermediate shade of skin colour, but it appears so because of presence of red and white hairs evenly present at the skin. It is clear from it that none of the two genes is dominant over the other.

Landsteiner and Levine discovered MN blood type in man on the basis of specific antigen present in RBC, produce specific antibodies. There are three phenotypes M, N and MN which are produced by $L^M$ and $L^N$ genes.

| Phenotype | Genotype | Antigen 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 all their children will be of MN

## Difference between incomplete and co-dominance

| Incomplete Dominance | Co-dominance |
|---|---|
| ➢ Phenomenon of inheritance where expression of both contrasting allele blend in heterozygous condition.<br>➢ New phenotype produce as a result of incomplete dominance.<br>➢ Quantitatively both express equally.<br>➢ e.g flower colour in 4o'clock plants | ➢ Phenomenon of inheritance where expression of both contrasting allele does not blend in heterozygous condition.<br>➢ New phenotype does not produce as a result of co-dominance.<br>➢ Quantitatively both express unequally.<br>➢ e.g hair colour in cow or MN blood group. |

## Multiple Alleles

A trait may have more than two alternative forms but Mendels only found two alternative forms of a gene. The genes which have more than two alternative forms are called multiple alleles. These multiple forms of gene are produced by gene mutation. These multiple forms of gene occupy same gene locus on chromosome, some traits have more than 100 alleles but each individual has two of them in diploid cell or only one in haploid cell. A well-known example of multiple allele in human being is ABO blood group.

Karl Landsteiner in 1901 discovered ABO blood group, has four different phenotype on the basis of presence of A or B antigen on the surface of RBC i.e a person with antigen A has a **blood group A** and that with antigen B has **blood group B**. A person with A and B antigen **blood group AB**. Similarly a person without both antigens on its RBC has **blood group 0**.

The genetic basis of ABO system was explained by Bernstein in 1924. He explained that there are three alleles responsible for the trait of blood group i.e. $I^A$, $I^B$ and i. These alleles have six possible combinations i.e. genotype, i.e. $I^AI^A$, $I^Ai$, $I^BI^B$, $I^Bi$, $I^AI^B$, ii.

**Table: 23.2 ABO blood groups of human**

| Blood Group | Genotype | Antigen | Phenotype |
|---|---|---|---|
| **Blood Group A** | $I^AI^A$ | A | A homozygous |
| | $I^Ai$ | A | heterozygous |
| **Blood Group B** | $I^BI^B$ | B | homozygous |
| | $I^Bi$ | B | heterozygous |
| **Blood Group AB** | $I^AI^B$ | A & B | AB heterozygous |
| **Blood Group O** | ii | No antigen | O homozygous |

## Co-Dominance in Human Blood Group AB

The phenomenon of Inheritance where both contrasting alleles of same gene locus express independently without affecting each other called co-dominance. In co-dominance the phenotype of both the alleles becomes apparent. Human AB blood group is the best example of co-Dominance, where both $I^A$ and $I^B$ are co-dominant alleles produce two different antigens on the surface of same red blood cells. Therefore the person with $I^A$ $I^B$ genotype have Blood group AB.

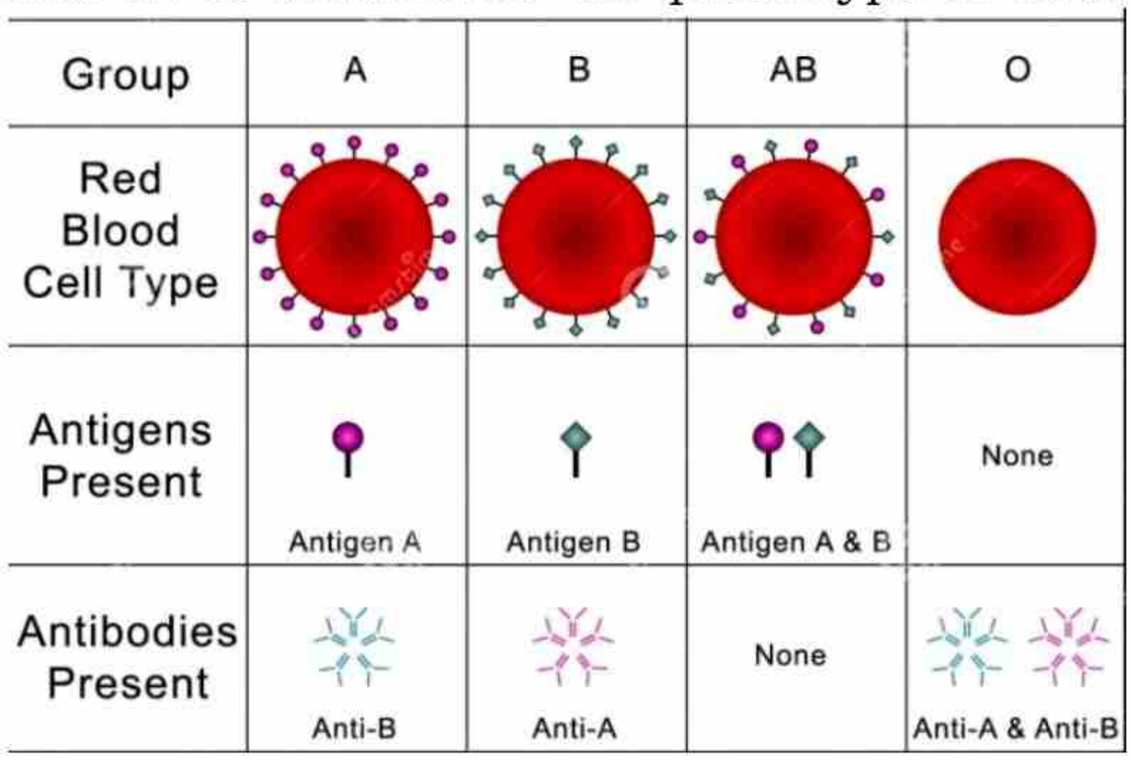



**Fig.23.3 ABO blood system**

## Human Blood Group System

In human beings numbers of different blood group systems are investigated to explain the variability of cells, chemical and its interaction with other blood groups. It is observed and carefully selected at the time of blood transfusion therefore International Society of blood transfusion has found more than 30 types of blood group system. These systems are mainly based on the presence or absence of special molecules found on blood cells. These molecules are mainly present on the surface of R.B.C and belong to the group of conjugated molecules i.e glycoprotein. The main type of blood group

systems are MN blood group system, ABO blood group system and $R^h$ system among these three, two are widely used i.e. ABO and $R^h$ system.

It is due to the reason that the incompatibility between donor and recipient lead to the death of recipient. The MN blood group system is considered as the rare blood group.

## Multiple Alleles of ABO Blood Group System

As we have discussed earlier that ABO blood group system is controlled by three alleles $I^A$, $I^B$ and i. I stand for **isoagglutinogen**, other term stand for isoagglutinogen is antigen: Allele $I^A$ specifies production of antigen A and allele $I^B$ produce antigen B but allele i does not produce any antigen. Both $I^A$ and $I^B$ are dominant allele whereas i is recessive by $I^A$ and $I^B$. These alleles start their expression at early embryonic stage and keep on expressing till death. Therefore the blood group of a person never changes throughout its life.

**Table: 23.2 Blood group systems in human based on R.B.Cs antigen.**

| | Blood group system | Antigen on surface protein |
|---|---|---|
| 1 | ABO | $A^1$, $A^2$, $A^3$, $A^4$ B and others |
| 2 | Rhesus ($R^h$) | D, C, c $C^w$ ,Cx, E, D4 and other |
| 3 | MNS | M, N, S, s |
| 4 | P | P, $P^1$ and $P^k$ |
| 5 | Lutheran | $Lu^a$, $Lu^b$ |
| 6 | Kell | K, k, $K^a$, $K^h$, $Js^a$, $Js^b$ |
| 7 | Lewis | $Le^a$, $Le^b$ |
| 8 | Duffy | $Fy^a$, $Fy^b$ |
| 9 | Diego | $Di^a$, $Di^b$ |
| 10 | Yt | $Yt^a$, $Yt^b$ |
| 11 | I | I, i |
| 12 | Xg | $Xg^a$ |
| 13 | Kidd | Kj |

The person having A Blood group produce antibodies called anti-B antibodies in its blood serum. while the person of B-Blood group produce Anti-A antibodies Person of AB blood group neither have anti-A nor have anti-B-antibodies in its Serum. The Serum of 0-blood group person has both anti-A and anti-B antibodies. The blood serum which contains anti-bodies called antiserum.

There antiserum appear in plasma during the first month after birth. The person with A blood group which contain Anti-B antibody if transfused with B-blood group will agglutinate or clump R.B.C of B-antigen (B-blood group) on the other hand R.B.C of B-blood group will agglutinate by A-blood group R.B.C, while R.B.C of AB blood group will not agglutinate by any of them and O blood group will agglutinate by any blood group. The blood transfusion is only safe if it does not agglutinate. Agglutination is dangerous for health of a person because Clumped cells form a huge mass which cannot pass through fine capillaries. The blood samples of the donor and the recipient are cross-matched for compatibility before transfusion. If wrong (non-compatible) transfused hemolytic reaction occurs which become fatal for the person. In hemolytic reaction, the antibodies of the recipient destroy the R.B.C of donor or the antibodies of the donor hemolytic the R.B.C. of the recipient.

The person have A and AB blood group receive blood of A-group, the person of B and AB blood group can receive blood of B-Blood group because they do not have anti-B antibodies. The persons of AB blood group can donate their blood only to the persons of AB-blood group because they have neither anti-A nor anti-B antibodies, while O blood group can donate to persons of all blood group because these persons produce both anti-A and B-antibodies but person of O-blood group can accept only blood of O-group because it has neither A nor B-antigen on its R.B-Cs.

## O-Ve as Universal Donor

If a person has O-ve blood group means it has neither antigen A, nor antigen B and antigen $R^h$ is also absent on R.B.Cs, The O produce anti-A and B antibodies, these antibodies are quickly absorbed by recipient tissues or greatly diluted in the recipient blood streams this do not develop agglutination of R.B.Cs. there **O-ve blood** is called **universal donor**. On the other hand side **AB +ve blood** group individuals are called **universal acceptor** because they can receive blood from the persons of all blood groups.

Some person also have A and B antigens in saliva and other body tissues, these persons are called secretors. They carry dominant secretor gene Se on chromosome number 19.

## $R^h$ Blood System

Usually when we tell our blood group, we mix-up two different systems of blood, which are controlled by two different genes, although the expression of both genes appear phenotypically at the surface of R.B.Cs, this $R^h$ blood system is usually represented by +ve or –ve sign, which refers to the presence or absence of $R^h$ antigen. $R^h$ blood group systems are explained on the basis of $R^h$ antigen present on the surface of R.B.C. This antigen observed in Rhesus monkey by Landsteiner in 1930s.

The prime gene locus D is mainly responsible for the production of and $R^h$ antigen the human population has two alleles of D i.e. **D** and **d**. The D is completely dominant over d. The person having DD, Dd genotype produce Rh antigen on R.B.C and called $R^h$ +ve while a person with dd genotype is unable to produce $R^h$ antigen therefore called -ve $R^h$ Unlike the naturally occurring anti-A and Anti-B antibodies of ABO system, anti-$R^h$ antibody production require a stimulus by human $R^h$ antigen itself. If a person of –ve $R^h$ receive blood of $R^h$ +ve person which carry $R^h$ antigen. The Recipient will begin to produce anti- $R^h$ antibodies against $R^h$ antigen or R.B.C of donor and start destroying it to from clump. On the other hand side -ve $R^h$ blood which does not contain any anti- $R^h$ antibody can be transfused to $R^h$ +ve recipient.

## Erythroblastosis Foetalis

It is a maternal foetal $R^h$ incompatibility problem. It occurs when Rh-ve women marry a $R^h$ +ve man, she conceive a child having $R^h$ +ve blood. If this $R^h$ +ve man has genotype DD, all of their children will have Dd genotype and phenotypically they are $R^h$+ve. On the other hand side if the genotype of man Dd (heterozygous) than the chances of $R^h$+ve and $R^h$ –ve in their children will be 50% both. The chance of Erythroblastosis foetalis is found when an $R^h$ +ve foetus is conceived by $R^h$–ve mother.

## Erythroblastosis foetalis an antigen-antibody Reaction

It starts when a $R^h$-ve mother conceive $R^h$+ve foetus. At the time of 1st labour some $R^h$ antigen seep into mother's blood which produces antibodies against it in the body of mother. At the time of next $R^h$+ve baby (foetus) some of the anti- $R^h$ antibodies seep through

placenta into the blood of $R^h$ +ve foetus and start break down of R.B.C i.e haemolysis. If this destruction remain continue the foetus become anemic, which start releasing immature erythroblasts in blood stream therefore this antigen-antibody reaction is called erythroblastosis foetalis. This anemia may lead to abortion or still birth or if pregnancy continues, the liver and spleen of foetus become enlarge due to fast destruction of R.B.Cs. The breakdown of R.B.C. produce large amount of bilirubin pigment which accumulate in the foetus and damage the neuron. It also tums skin and white part of eyes into yellow this problem is called jaundice. It means if the baby remained alive in this condition suffers from severe hemolytic anemia and jaundice.

## Measures to counter the problem of Erythroblastosis foetalis

The $R^h$-ve mother is given $R^h$ antiserum in injectable form during early pregnancy and immediately after labour. The $R^h$ antibodies in $R^h$ antiserum will destroy $R^h$ R.B.C of the foetus before they stimulate production of maternal anti-$R^h$ antibodies. The injected antiserum vanishes before next pregnancy therefore $R^h$ antiserum should be given at the time of each pregnancy. If a baby is born with this defect, the blood with high level of bilirubin of baby should be immediately replaced by $R^h$ negative blood free from anti-$R^h$ antibodies

## Polygenic Inheritance and Epistasis

The traits are expressed either qualitatively or quantitatively. We have discussed qualitative inheritance in our pervious discussion where traits are controlled by one gene present on single gene locus e.g. Tall, dwarf, blood group, $R^h$ factors etc. Although these gene have multiple alleles, the phenotype was dependent on the presence of a particular allele. On the other hand some traits are controlled by many genes located at different loci.

Their phenotype will work in the additive manner therefore, this inheritance is quantitative inheritance. The phenomenon of inheritance where a single trait is controlled by two or more than two separate pairs of genes which manifest themselves in additive manner to yield variable phenotype called Polygenic Inheritance and these traits are called continuously varying traits.

On contrary there is another phenomenon of inheritance where expression of a gene is controlled by another gene which is located on different locus. This phenomenon of interference in called Epistasis (Gr- Epi- above, stasis = standing or stop). In this phenomenon, non-locus genes alter the expression of original gene which is independently inherited. It should not be confused with dominance because dominance is the relationship between allele of same gene located at same gene locus but epistasis is the interaction between different genes, located at different gene loci.

## Polygenic Traits in Plants and Animals

As we have discussed above that a continuously varying trait is encoded by alleles of two different gene pairs located at different loci, all work for a same trait and express in additive manner to show quantitative effect, are called polygenes. Each gene of a polygenic group has a small positive or negative effect on the character. Polygenes support each other to produce positive or negative effects e.g wheat grain colour in plant and skin colour in animals.

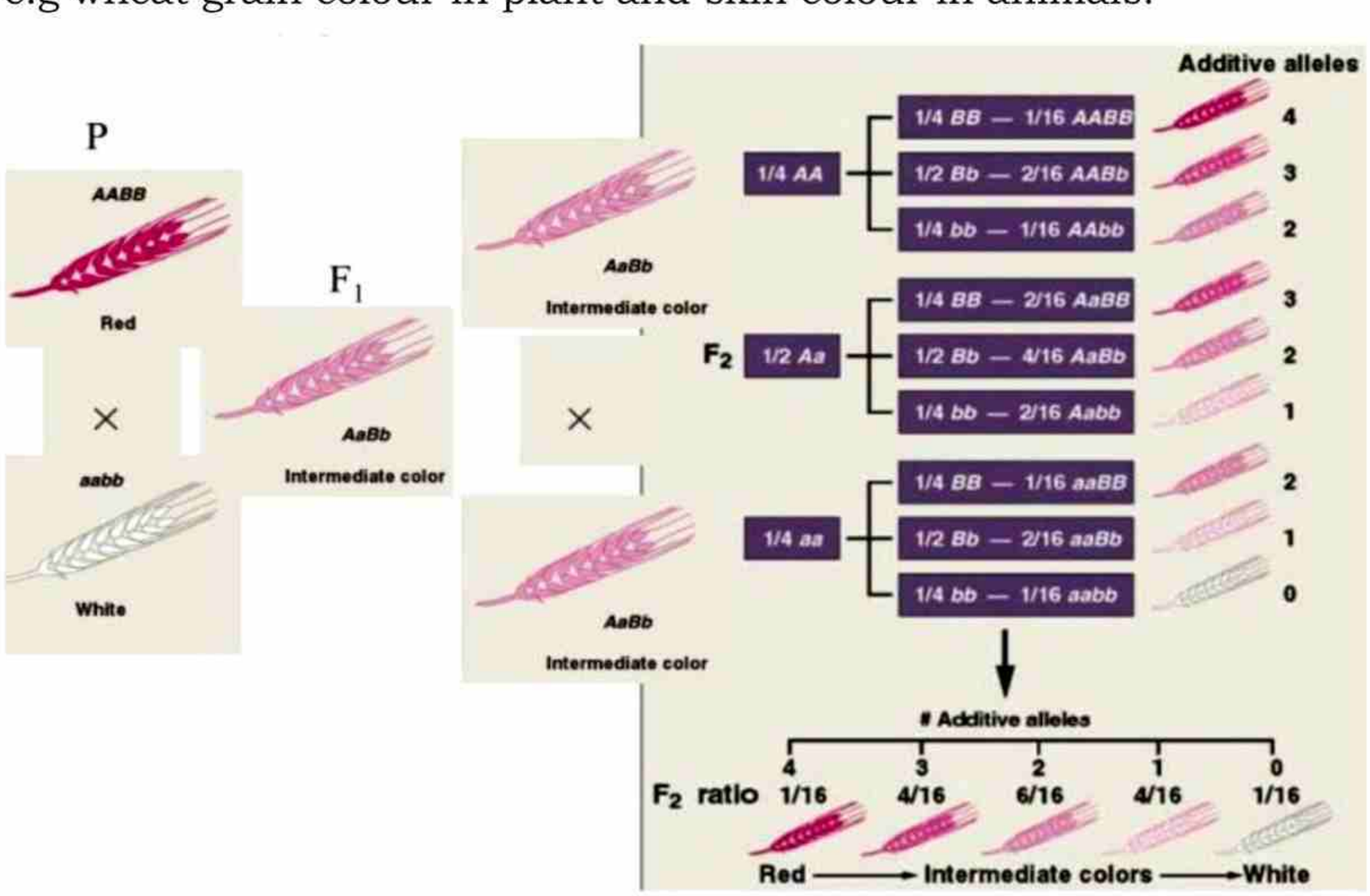



**Fig. 23.4 Polygenic trait of wheat**

## Wheat grains colour trait as polygenic trait

The colour of wheat grains vary from white to dark red. It is a best example of polygenic inheritance or continuously varying trait. Usually seven different phenotypes are found in the population of wheat all over the world. These phenotype vary from dark red to white.

Nilsson-Ehle studied the genetics of wheat grain colour. When he crossed true breeding dark red with white grain producing wheat plants, All plant in $F_1$ generation produce light red grains. i.e intermediate between two parental shades. It seems like case of incomplete dominance, so when $F_1$ grains were grown to mature plants and crossed with each other, $F_2$ grains had exactly seven shades of colour in the ratio of 1 dark red: 6 moderately dark red: 15 red: 20 light red: 15 pink: 6 light pink: 1 white as given in chart below.

Phenotypes In wheat grain colours

| Phenotype | Dark red | Moderately dark red | Red | Light red | Pink | Light pink | White |
|---|---|---|---|---|---|---|---|
| Ratio | 1 | 6 | 15 | 20 | 15 | 6 | 1 |

It was found that the grain is controlled by three different gene pairs. i.e Aa, Bb, Cc at three different loci, Each individual would contain six alleles for this trait. Allele A, B, C are for the production of red pigment while a, b, c does not produce any pigment. If a wheat plant contains ABC in the combination of **AABBCC** all produce red pigment so they produce Dark red grains. If the genotype will be **aabbcc** none of them produce red colour so the wheat grain colour will be white. The other genotype will be developed different colours of grains as phenotype.

| | ABC | ABc | AbC | Abc | aBC | abC | aBc | abc |
|---|---|---|---|---|---|---|---|---|
| ABC | AABBCC | AABBCc | AABbCC | AABbCc | AaBBCC | AaBbCC | AaBBCc | AaBbCc |
| ABc | AABBCc | AABBcc | AABbCc | AABbcc | AaBBCc | AaBbCc | AaBBcc | AaBbcc |
| AbC | AABbCc | AABbCc | AAbbCC | AAbbCc | AaBbCC | AabbCC | AaBbCc | AabbCc |
| Abc | AaBbCC | AABbcc | AAbbCc | AAbbcc | AaBbCC | AabbCc | AaBbcc | Aabbcc |
| aBC | AaBBCC | AaBBCc | AaBbCC | AaBbCc | aaBBCC | aaBbCC | aaBBCc | aaBbCc |
| abC | AaBbCC | AaBbCc | AabbCC | AabbCc | aaBbCC | aabbCC | aaBBCc | aabbCc |
| aBc | AaBBCc | AaBBcc | AaBbCc | AaBbcc | aaBBCc | aaBbCc | aaBBcc | aaBbcc |
| Abc | AaBBCc | AABbcc | AAbbCc | AAbbcc | AaBbCc | AabbCc | AaBbcc | aabbcc |

It was found that all the six allele code for red pigment (AABBCC), colour of grain will be dark red. When all six alleles do not

produce red (aabbcc) the colour of grain will be white. When a grain has one allele for red light (Aabbcc or aaBbcc or aabbCc) the colour of gram will be light pink, if plant contain two alleles of red pigment (AaBbcc or aaBbCc or AabbCc) the colour of grain will be pink. If the plant has three Pigments of Red colour (AaBbCc or AABbcc or AabbCC) will be light red. In the same way for alleles colour dose (AABBCC & aaBBCC or AAbbCC) will express red and five dominant alleles dose (AABBCc or AABbCc or AaBBCC) will produce moderately dark red grains. It showed that the phenotype of grain colour depend upon the presence of colour producing alleles i.e. A, B and C. The expression of these genes are also influenced by environmental factors like light, Water and nutrients.

## Inheritance of Human Skin Colour

Another example of polygenic trait is human skin colour trait, which is found in variable shade therefore it is continuously varying trait. Human skin colour develops by the presence of a pigment melanin. If a person produces high quantity of melanin, its complexion (skin colour) will be dark or vice versa. This trait is controlled by three to six gene pairs, greater the number of melanin producing gene darker will be the skin colour. These genes are working in the following manner:

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| Locus 1 | $d^1d^1$ | $d^1D^1$ | $d^1D^1$ | $D^1D^1$ | $D^1d^1$ | $D^1d^1$ | $D^1D^1$ |
| Locus 2 | $d^2d^2$ | $d^2d^2$ | $d^2D^2$ | $D^2d^2$ | $D^2d^2$ | $D^2D^2$ | $D^2D^2$ |
| Locus 3 | $d^3d^3$ | $d^3d^3$ | $d^3d^3$ | $d^3d^3$ | $D^3D^3$ | $D^3D^3$ | $D^3D^3$ |
| Total number of dark-skin genes | 0 | 1 | 2 | 3 | 4 | 5 | 6 |


**Fig.23.5 Polygenic trait of human**

**Gene A** is responsible for permanent survival, proliferation and migration of melanocytes

**Gene B** is responsible to synthesize tyrosinase, involve in the conversion of tyrosine into melanin. It means that it is involved in the synthesis of melanin.

**Gene C** is responsible for determination of either proper black or dark brown type of melanin i.e. eumelanin will synthesize or red-brown type of melanin i.e. phenomelanin will produce.

Each of the above ABC has & two forms, the dark skin alleles are represented by capital letters (ABC) and light skin alleles are represented by small letters (abc). These alleles are not completely dominant over each other. In heterozygous condition they exhibit intermediate phenotype i.e. incomplete dominance, the gene a, b and c act as light-skin alleles in the genotype they inhibit melanin production.

There are seven different shades of skin colour ranging from very Light (aabbcc) to very dark (AABBCC), mostly human beings have the intermediate skin colour (AaBbCc) this genotype develops mulato i.e. an offspring of intermediate skin colour belongs to black and white parent. When individual of mulato cross with each other i.e. AaBbCc x AaBb Cc, each parent can produce eight different types of gametes, when these gametes fertilize there will 64 chances of zygote formation by which offspring of seven colours can be produced.

## Epistasis

We have already discussed that the effect of non-locus on other gene to interfere in their phenotype is epistasis. The gene which interferes and masks the phenotype of the non-locus gene is called epistatic gene or interfering gene and the gene which expression masked by epistatic gene is called hypostatic gene.

## Examples of Epistasis in Plants and Animals

### Colour pigments trait in Foxgloves Petals

In foxgloves plant petal colour is determined by three genes. M which expresses to Synthesis an enzyme to develop Anthocyanin which expresses as purple pigment, mm produces no pigment it seen albino with yellowish spot on petal, while another gene D is an

enhancer of anthocyanin due to this gene expression more anthocyanin produce and petal become darker, dd does not enhance. The third gene which is located at another gene locus, W prevent pigment deposition except in the spot as a result white spotted petals are produced whenever ww allow pigment deposition in petals.

M (MM, Mm) → Purple
mm →white (no pigment)
W (WW, Ww) → spots

D (DD, Dd) → enhancer of purple
dd → no enhancer (light)
ww → Uniform colour (no-spot)

According to the result of di-hybrid cross performed by Mendel if genes of 2 traits are heterozygous in both parents they produce offspring with the phenotype ratio of 9:3:3:1 but there was no epistatic genes, but In the case of foxgloves plant di-hybrid cross where both are heterozygous for both traits. Produce 12: 3:1

DdWw x DdWw

Gametes DW Dw dW dw x DW Dw dW dw

| | DW | Dw | dW | dw |
|---|---|---|---|---|
| DW | DDWW White spotted | DDWw White spotted | DdWW White spotted | DdWw White spotted |
| Dw | DDWw White spotted | DDww Dark red | DdWw White spotted | Ddww Dark red |
| dW | DdWW White spotted | DdWw White spotted | ddWW White spotted | ddWw White spotted |
| dw | DdWw White spotted | Ddww Dark red | ddWw White spotted | Ddww light red |

Dominant Epistasis: white spotted : Dark red : light red

12 : 3 : 1

The mechanism of interference shows that **W** prevents deposition of pigment in flower, only in spots where as **w** allows deposition in the all cells of petals. On the other hand **D** allele allows to synthesize large amount of anthocyanin (red pigment whereas allele **d** synthesize little amount of anthocyanin.

## Coat Colour in the Labrador retriever
## Example of Epistasis in Animal

Labrador is an excellent type of dog its coat colour is one of the best example epistasis in mammals. These dogs are found in three colours due to their coat colour, i.e. Black, chocolate and yellow. This coat colour Inheritance is autosomal i.e. their genes are located at autosome it means it is not related with their sexes: the **B** is for Black colour **b** is for Chocolate colour both are located at same gene locus but the gene of yellow colour **E** is located at other gene locus.

The Black Coat colour (B) is dominant over chocolate (b). The Chocolate puppy must be homozygous i.e. both parent donates recessive allele. Its genotype well be (bb) but if any puppy have BB in Bb genotype to offspring will always be black, yellow coat colour gene is located at different gene locus this gene also have two alleles **E** dominant **e** recessive produce yellow it requires two doses of ee it means homozygous recessive will interfere in the expression of Black and chocolate to produce yellow coat colour. It shows that the colour gene is recessive epistatic gene. However, homozygous dominant EE or heterozygous dominant will not interfere to the puppies will be either black or chocolate according to parental gene combination. If a Black Labrador which is homozygous for both gene pair (BBEE) is crossed with a yellow (bbee), all offspring will be black with BbEe genotype. If these black offspring interbreed produce offspring of all three coat colours i.e. Black, Chocolate and yellow.

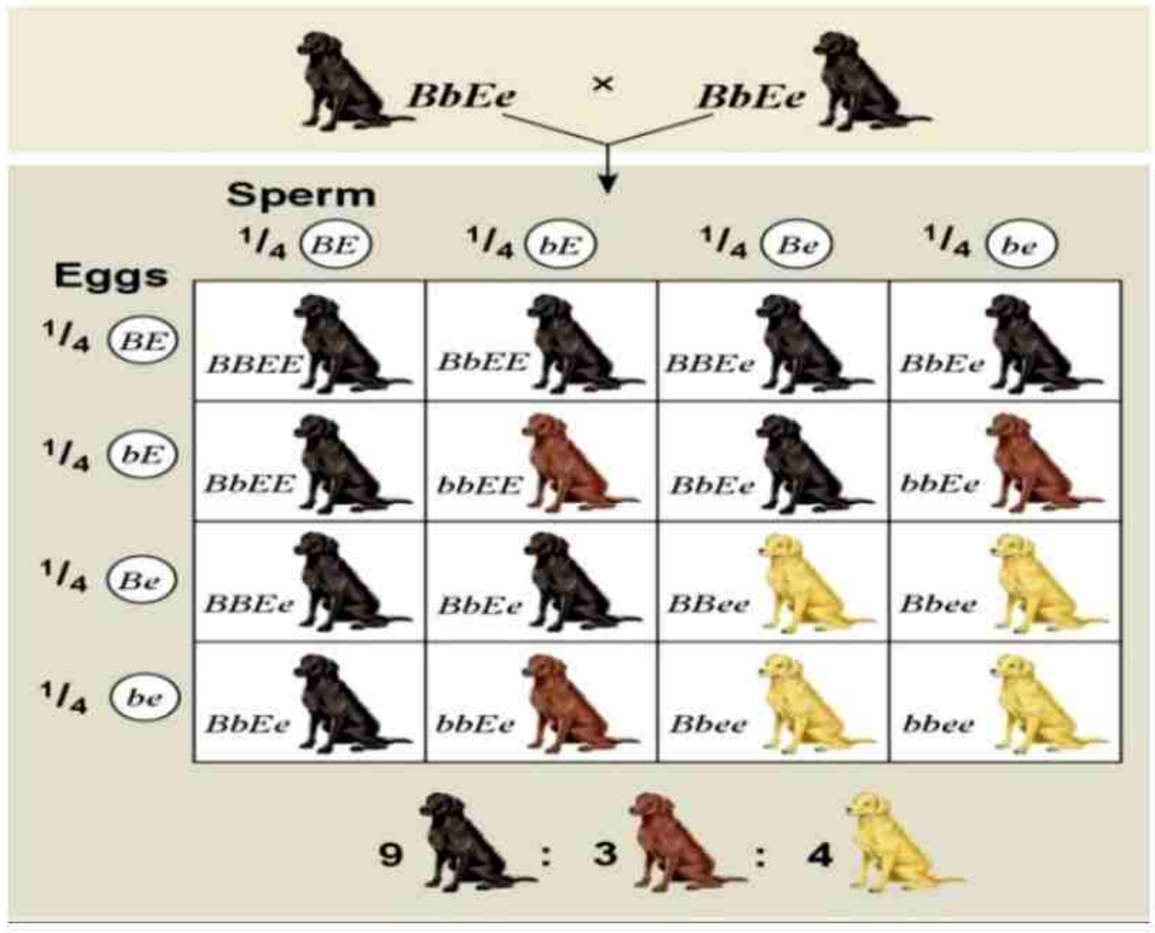



**Fig.23.6 Dominant Epistasis**

## Gene Linkage and Crossing Over

According to careful estimates, there are thousands of genes located on four (04) pairs of Chromosomes present in the cells of Drosophila. It means that each chromosome contain a large number of genes, similarly chromosome of all organism have many genes, located at single chromosome. It was found that a chromosome may contain maximally 4000 genes. According to chromosomal theory of inheritance each genes are inherited by chromosomes. It means that genes located at a single chromosome tend remain together during inheritance. This tendency of gene in a chromosome to remain together is called **Linkage** and the genes which are present at some chromosome and inherit together called **Linked gene**, the genes, which are located on autosome called autosomal linked genes and their inheritance called **autosomal inheritance** or **autosomal linkage**. Similarly if they are located on sex chromosome (X or Y) their linkage called **sex-linkage** and their inheritance is called **sex-linked inheritance**. All the linked genes present on the same chromosome at the time of inheritance called **linkage group**. therefore the number off linkage groups in an organism are equal to number of homologous chromosomes present in an organism.

The linked genes are present on same chromosomes therefore they are considered to be inherited together in the offspring so always parental combination of characters are found in offspring and assorted traits do not segregate independently. Linkage between genes can be detected by test cross of di-hybrid organisms. It is a cross where a heterozygous organism of two traits is **back crossed** with its recessive parent. If the results will come in the following ratio, then the linkage will be determined easily i.e. if four phenotypic combinations (parental and non-parental) occur in offspring then there will be no linkage between the genes. If this ratio is deviated and more parental types with less non-parental type appear which shows **partial linkage**, if only parental type appears it shows **complete linkage**. In Mendelian di-hybrid cross, where linkage was not considered as phenomenon the result shows the parental and non-parental combination is the ratio of 9:3:3: 1. In case of linked genes of di-hybrid cross the result will be only parental combination with a ratio of 3:1.

T.H. Morgan was the geneticist who observed karyotype of Drosophila, he was of the contributor of **Chromosomal theory of Inheritance**, also studied 85 pairs of contrasting trait inheritance in *Drosophila melanogaster*. In Drosophila he observed two assorted traits whose genes are linked together (that is located at same chromosome). The dominant gene **V** for normal wings and its recessive allele **v** for vestigial wings, another trait where a dominant gene **B** for Gray body colour is dominant over black colour gene (**b**), Both are located at same chromosome.

As they are linked, they tend to be inherited together i-e. **V** with **B** and **v** with **b**. It implies that when a homozygous **VVBB** fly is crossed with a homozygous **vvbb** fly all offspring will have **VvBb** i.e. Normal wings and Gray body colour. When this heterozygous fly is crossed with homozygous recessive **vvbb** and the genes remains completely linked as well as no crossing over occur then only two parental type in offspring will appear equally i.e. Gray body with normal wings and black with vestigial winged.

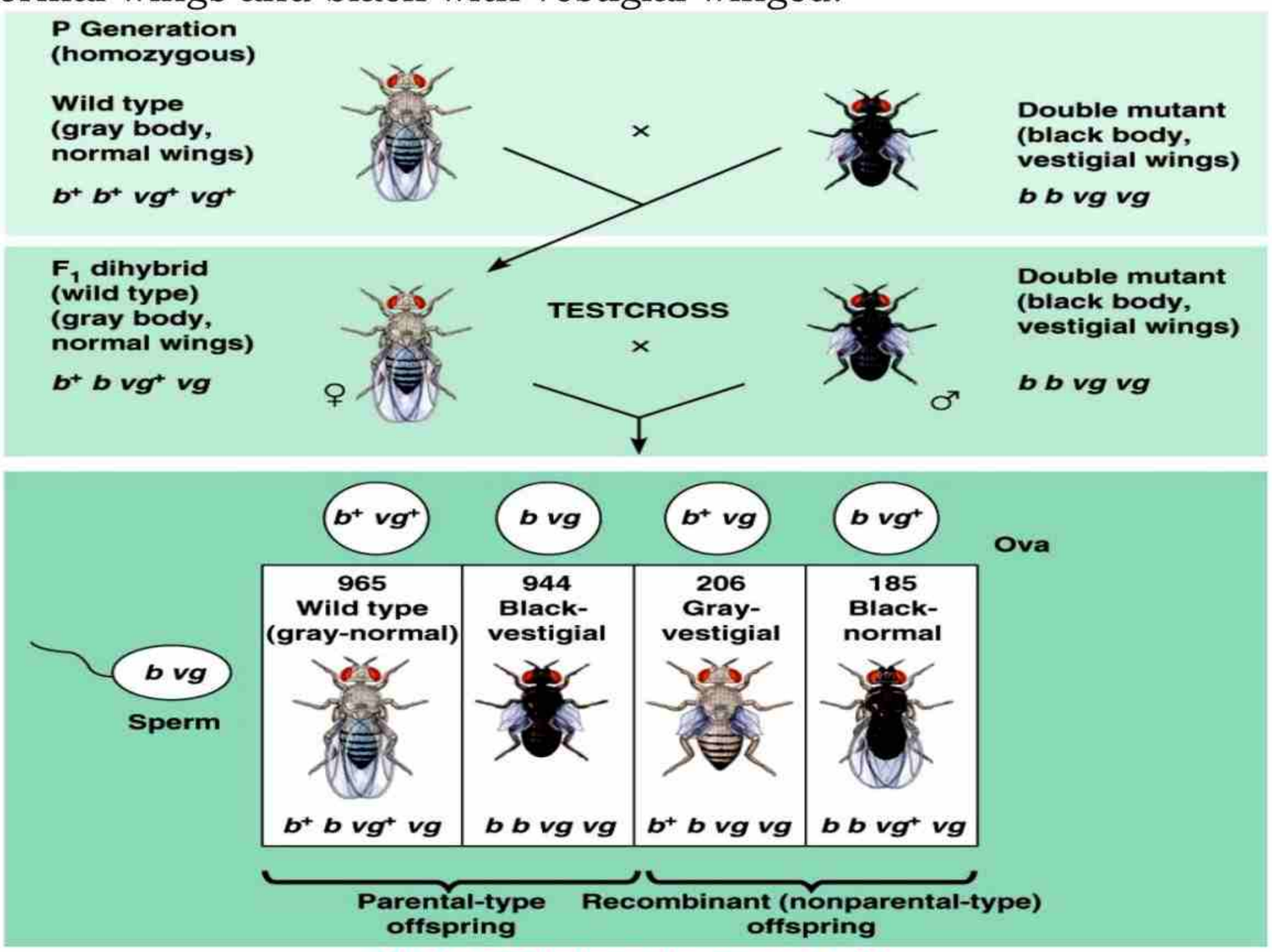



**Fig.23.7 Linkage in Drosophila**

## Crossing Over

The linkage is also not an absolute and it not necessary the genes of a chromosome remain attached with each other and transmits together. If Linkage remain continue so the inheritance of a trait remain continue and the offspring will be like one of the parent only. The gametes in animals and spore mother cells (S.M.Cs) divide by meiosis to produce gametes, and spores respectively. During meiosis, the homologous chromosomes pair up, this pairing of homologous chromosomes called **Synapsis**. Soon after meiosis they attach and form cross bridges between their non-sister chromatids, they sometimes exchange their segments, this process of exchange of chromosomal segment between non- sister chromatids of homologous Chromosome is called **crossing over.**

After separation the chromosomes carry some genes with each other and new combination of gene develops on a chromosome which develops variety of gene and new combination between parental combinations occurs. The recombinant Chromatids resulting from crossing over may bring alleles together in a new combinations so, when gametes, are formed gametes a variety in gametes develop. Therefore this process leads to genetic variability during sexual reproduction. Take an example to understand the process of crossing over, where a pair of homologous chromosomes carries gene **T** and **B** on both chromosomes where as another gene homologous has **t** and **b**. **Chiasmata** is formed between their non-sister chromatids and crossing over also takes place as alleles of non-sister chromatids are different, so exchange between their segment cause recombination of their alleles.

Allele **b** cross over to chromosome containing allele **T** and allele **B** cross over to chromosome containing allele **t**. These chiasmata open and sister chromatids also separate from each other to become an independent chromosome. At the time of gametes formation 4 types of gametes are formed two with parent combination i.e. **TB** and **tb** and two with non-parental (recombinant) combination i.e. **Tb** and **tB**. If crossing over will not occur only two parental types of gametes formed. The parental types of gametes produce offspring, with parental combination of characters while non-parental (recombinant)

combinations produce offspring with non-parental (recombinant) characters.

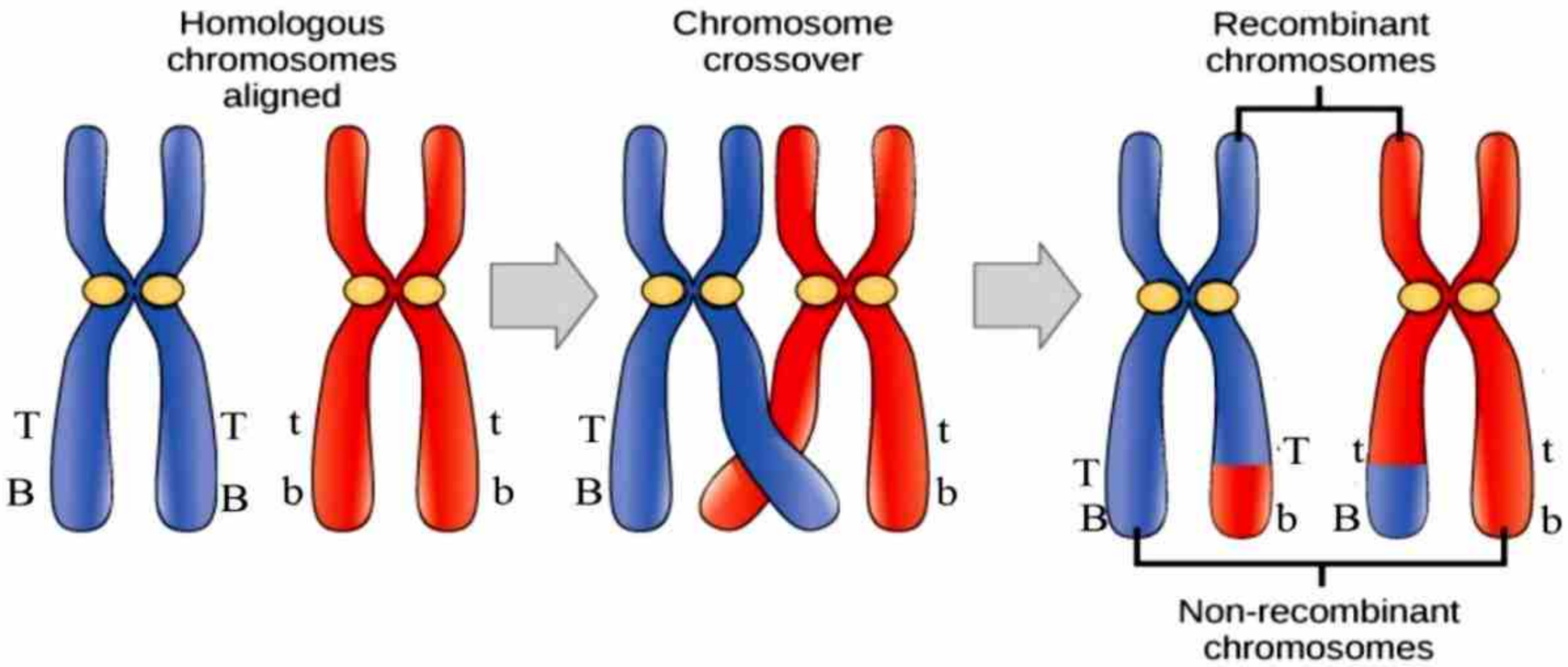



**Fig.23.8 Crossing over**

## Sex Determination

It was found in early year of last century that in the animals and plants most of the chromosomes are found in the form of homologous pair but a pair of chromosome may or may not be homologous in the members of same species. W. Sutton found the simplest situation of chromosomal difference in Grass-hoppers, where he found male have one chromosome less than female i.e. the females have 24 and male have only 23. It means the female has 12 pairs of chromosomes in their Somatic cell (2n) whereas 12 chromosomes (n) in each egg.

On the other hand male of Grasshopper have 11 pairs with a single chromosome in somatic cell (2n) whereas by spermatogenesis produces two types of sperms one type have 11 chromosomes (n) and other with 12 chromosomes (n). Fertilization of egg (12 chromosomes) have two chance, either with sperm having 11 chromosomes (n), or with the sperm have 12 chromosomes (n). If a sperm with 11 chromosomes fertilizes the egg, a male baby will develop or the other sperm (12 chromosomes develop female baby on fertilization this odd chromosome, which determines the sex of the individual is formed as **sex-chromosome**. The other chromosomes which are similar in male and females are **autosomes**.

## XX and XY sex-determination in Drosophila and Mammals

### Sex- Determination Drosophila

T.H. Morgan, a Nobel Prize winner (1933) selected. *Drosophila melanogaster* (Black-bellied dew lover), common fruit fly, can be seen hovering around over ripe fruit, for his experimental work. He observed the karyotype of this fly and noted that the male and female Drosophila has differences in one pair of chromosomes as shown in following Fig. 23.8

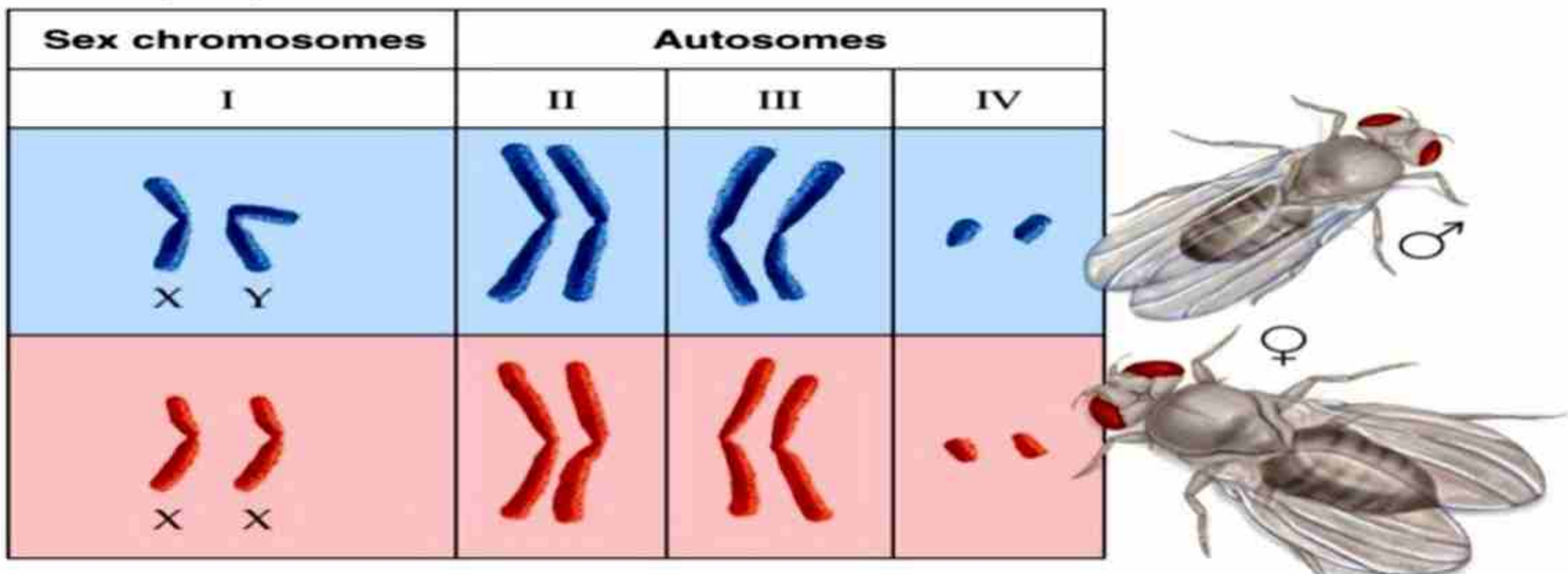



**Fig. 23.9 Karyotype of Drosophila**

This shows that there are three pairs of chromosomes which are same in male as well as in female fly. These chromosomes are called **autosomes** but the difference lies in 4th pair. The female has both the chromosome of 4th pair similar and rod shaped (Homologous). On the other hand male has both the chromosomes different from each other (Heterologous) one chromosome is rod shaped and other is hook shaped. This 4th pair designated as **sex-chromosome** which decide the sex in Drosophila.

Moreover, the rod shaped sex chromosome, two of the female and one of the male are alike designated as **X-chromosome**. The hook shaped unlike sex-chromosomes is designated as **Y chromosomes**. So, In Drosophila, individual having **xx** will be a female and that receive **xy** will be a male

### Sex-Determination in Human

In human beings and majority of animals, the case is same as Drosophila. The difference in male and female is not of one whole chromosome as Grass-hopper, but of the shape of one chromosome in one sex.

In man each cell either somatic or germ cell has 46 chromosome (2n) of which 44 i.e 22 pairs are similar in both sexes called **autosomes**. The 23rd pair differ in female consist of similar chromosomes (homologous) but in male the 23rd pair differ in shape, one of the two chromosomes is like sex-chromosomes of female of but the second is much smaller than the other. The two chromosomes of female's 23rd pair and one of male which are similar are called **X-Chromosome**, while the unlike, smaller chromosome of male's 23rd pair is called **Y chromosome**. So, the human female possesses 44 + XX chromosomes, whereas the male is 44+ XY chromosomes in their Karyotype.

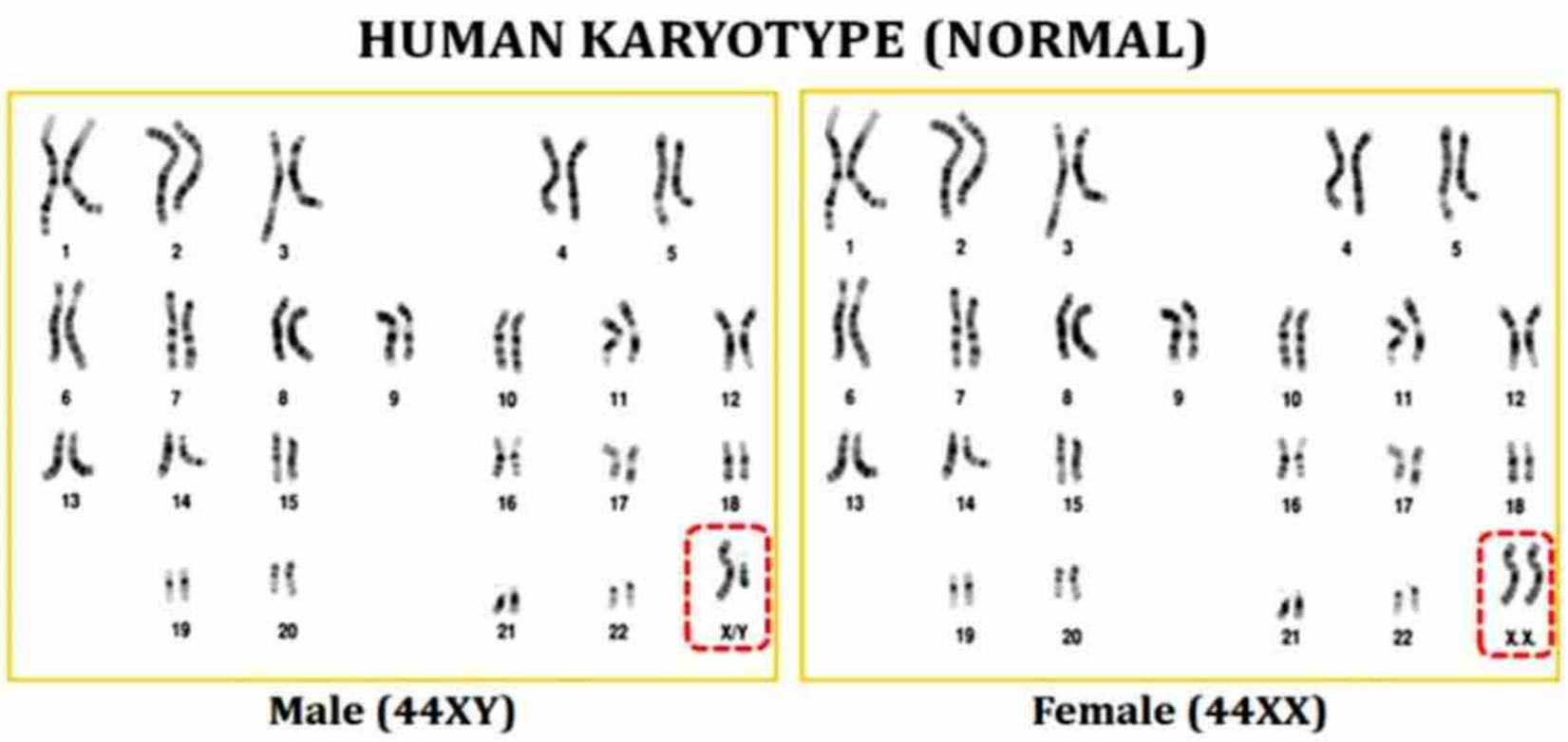



**Fig.23.10 Karyotype of human male and female**

The above stated situation that the male is **heterogametic** i.e. produces two types of sperms i.e. X and Y containing and the female is **homogametic** i.e. produce only one type of ovum containing only X.

## Other Patterns of Sex Determination

### (i) XO-XX Type

As we have discussed that in some animal only one chromosome work as sex-chromosome, and this pattern on sex-determination was found in Grass-hopper. The protenor bug also has same where male has only one sex-chromosome i.e XO. This sex chromosome is X-chromosome and other sex-chromosome is missing entirely in male. Therefore male of protenor bug is heterogametic i.e.

produce two types of sperms one type of sperm has X Chromosome while the other type do not contain sex- Chromosome. The gamete without sex chromosome is called Nullo gamete. Female of this bug have one X chromosome in all ovum therefore it is called homogametic, so sex of the offspring depend on the kind of sperm that fertilizes to ovum. If an x-carrying sperm fertilize the baby will be female, if the nullo-sperm fertilize the ovum the zygote will carry XO, so a male baby will develop from this zygote.

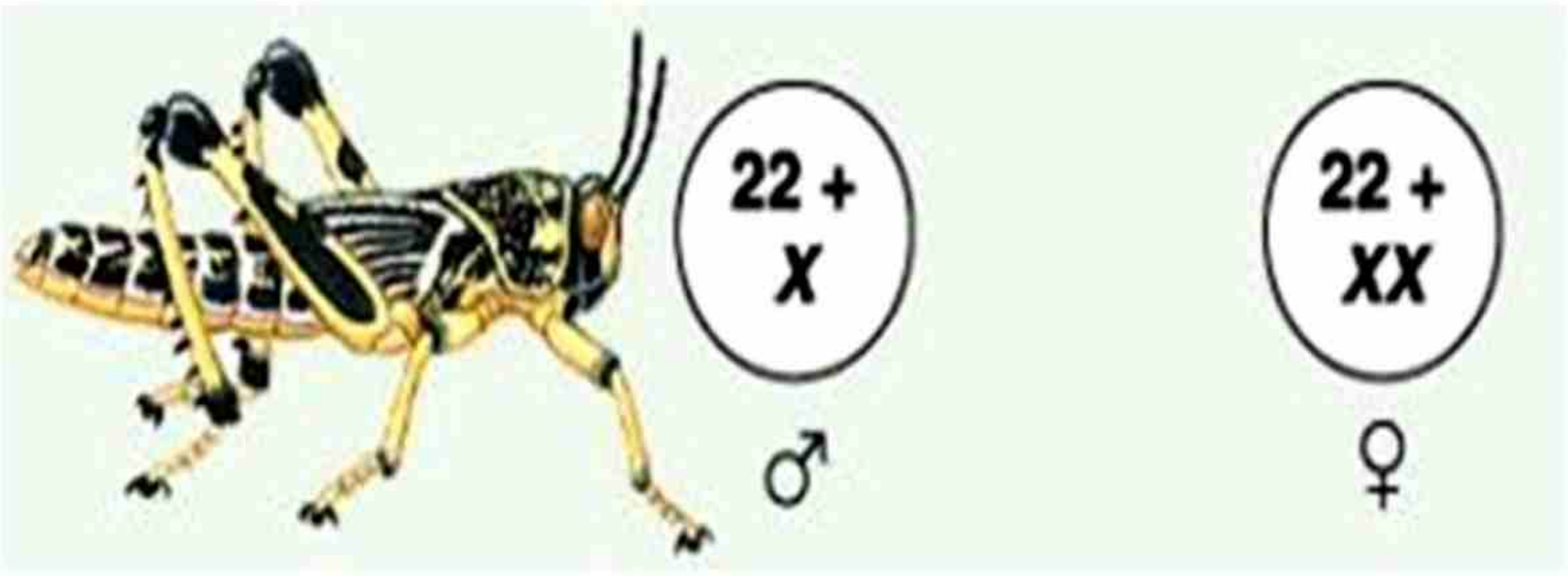



**Fig.23.11 Karyotype of grasshopper**

### ii) ZZ-ZW Type

In some organisms like birds, butter fly and moths the conditions of sex-determination is different from the above. J. Seiler in 1914 discovered that in moths the reverse of XX-XY System of sex-determination is present. In this system the female is heterogametic because she contains heterologous sex-chromosome therefore she produce two types of eggs some contain Z as sex chromosome and some contain W as sex chromosome in equal proportion.

On the other side all male produce same type of gametes have Z chromosome as sex-chromosome. It shows that the female which produce two variety of ovum become responsible for sex-determination in offspring, when a Z chromosome carrying egg is fertilized by sperm which always contain Z, male baby will produce but when a W carrying egg is fertilized by sperm, a female offspring is produced.

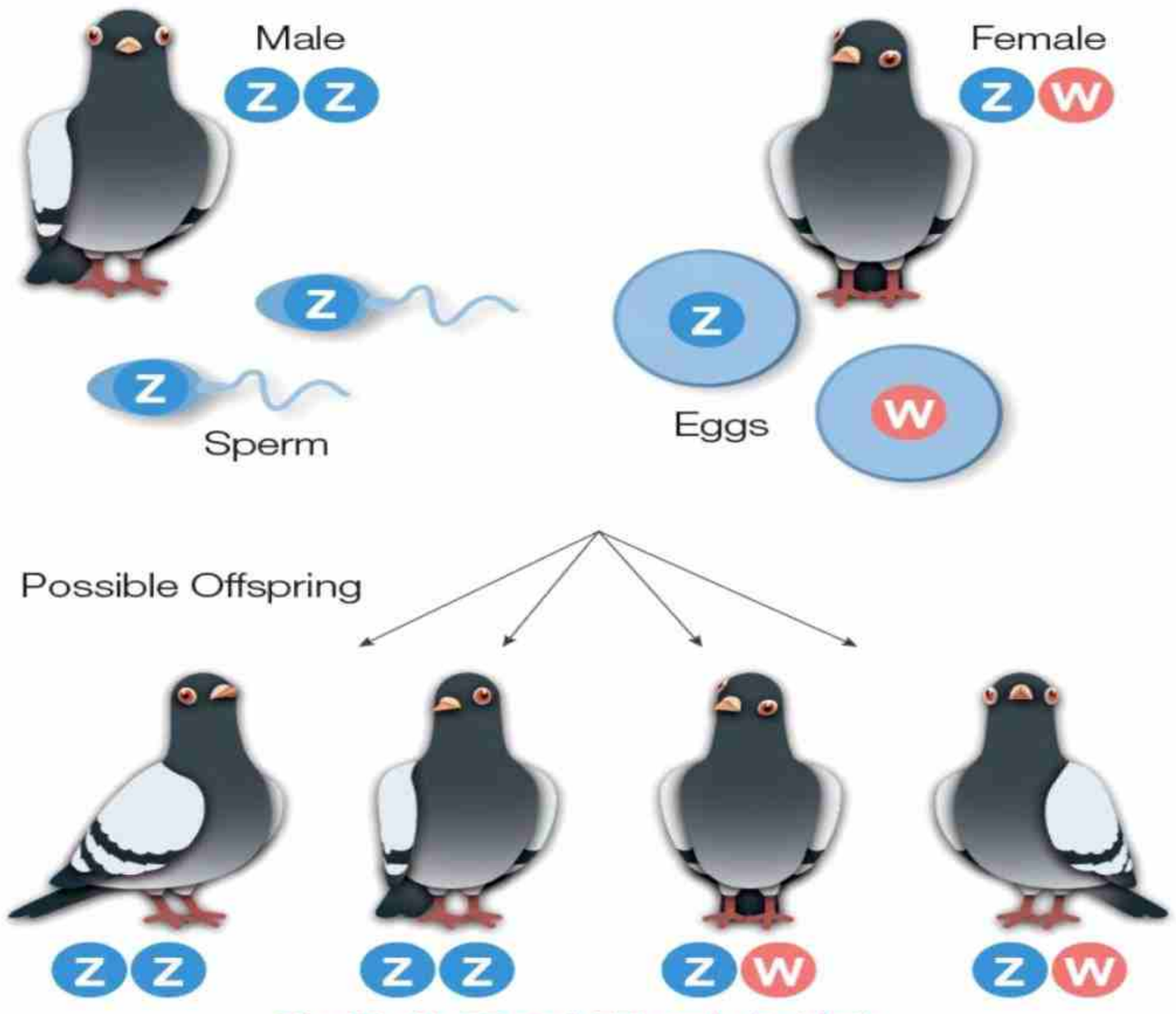



**Fig.23.12 ZZ and ZW trait in birds**

| | Male baby | | Female baby |
|---|---|---|---|
| Sex ratio | 2 | : | 2 |

**Genetic problem**

1. Draw a pedigree of 3 generations in a human family to show how many offspring will be male and female?
2. A moth produce four babies, how many male and female babies will produce in their next generation show it by develop pedigree.

## Sex- Linkage

According to chromosomal theory of inheritance, the genes are located at chromosome. The genes are located at same chromosome called linked genes because they usually inherit together in a linked manner if crossing over does not take place. The genes which are located at autosomes called autosomal linked gene and they inherit

through autosomes from parents to offspring. This type of inheritance called autosomal linked inheritance. On the other hand, the genes which are located at sex-chromosome are called sex-linked genes. The sex-chromosome in human or Drosophila are of two types X and Y. The alleles which are located at X-chromosomes called X-linked and in the characters which are inherited through the X-chromosome are called **X-linked inheritance** whereas the alleles located at Y-chromosome called Y- linked, and their inheritance called **Y-linked inheritance**. Some traits are controlled by that type of genes which have alleles found on both X and Y, such traits are called **X-Y linked** or **Pseudo autosomal** because they behave differently from sex-linked traits they have just autosomal mode of inheritance. On the other hand Y chromosome is not completely inert. It does carry a few genes which have no contrasting alleles on X-chromosome. The genes on Y chromosomes of human express characters of maleness e.g. bobbed gene of Drosophila are pseudo autosomal because they have pattern of inheritance like autosomal genes.

## Sex-Linked Inheritance in Drosophila

T. H. Morgan (1910) performed various breeding experiments with wild type red-eyed Drosophila. He noticed a white eyed mutant. This was male and turned out to be a true breeding strain of white eyed flies. He crossed this white eyed male with homozygous red-eyed female.

The $F_1$ and $F_2$ population followed the simple Mendelian ratio. But when white eyed female was crossed with red-eyed male, the result

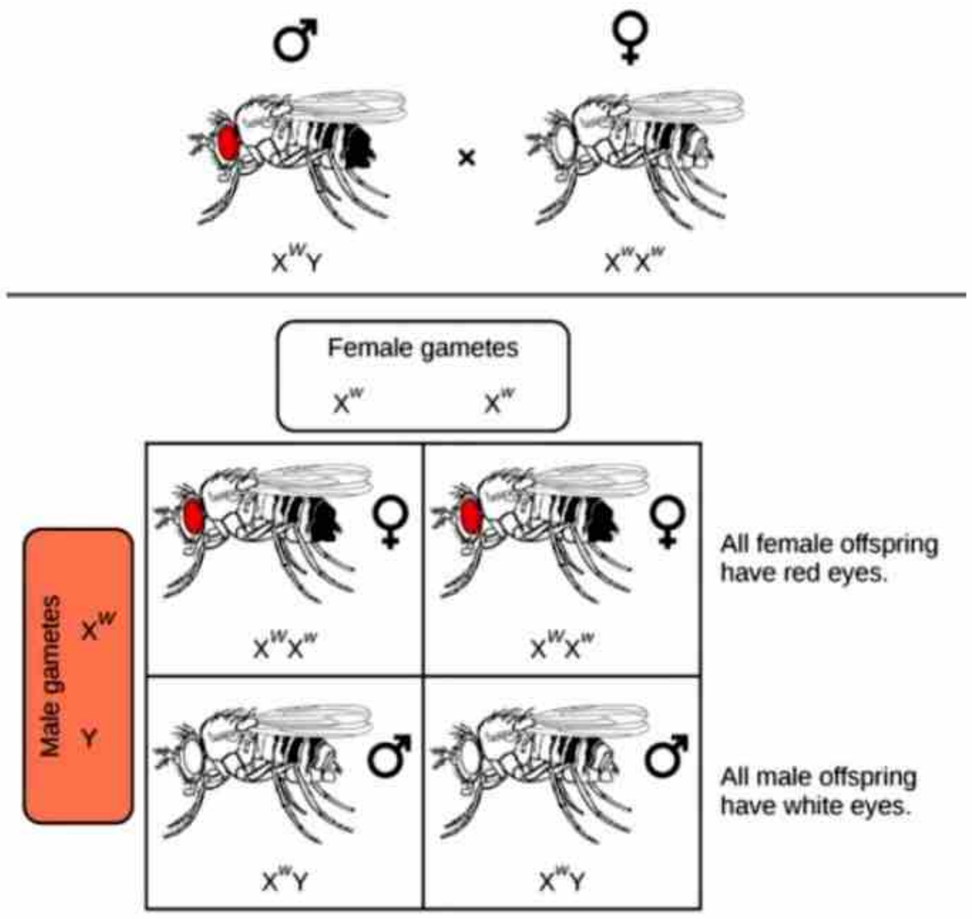



**Fig.23.13 Sex linked trait in Drosophila**

was not same as before. To examine the details of various cross-made by T. H. Morgan. Here the case in represented in the form of figures of Cross, where R for red-eyed and r for recessive white eyed flies.

### Cross- I

When red-eyed ($X^RX^R$) female is crossed with white eyed flies ($X^rY$) male the $F_1$ generation shows all red flies, the case in male and female are same.

The $F_2$ generation shows red-eyed and white eyed flies in ratio of 3:1. (All females are red but the chances of red eyed and white eyed type in male are 50% : 50%). In the some case genotypically females are also of two types, although both are red eyed.

The one type of female is homozygous i.e. unable to produce white eyed offspring while the other type of female are heterozygous (carrier for white eyed genes) i.e. can produce white eyed offspring

| Cross-I | | | Cross-II | | |
|---|---|---|---|---|---|
| Red eyed Female | X | white eyed Male | white eyed Female | X | Red eyed Male |
| $X^RX^R$ | | $X^rY$ | $X^rX^r$ | | $X^RY$ |

### Cross-II

When Red eyed male ($X^RY$) crossed with white eyed female ($X^rX^r$) the result were found different from cross I. In $F_1$ both types of flies were produced that is red eyed and white-eyed. Moreover all the red-eyed were female, while all the white eyed were males. In $F_2$ generation again red and white eyed appeared in equal ratio where half of them male and females are red as well as white eyed.

On the basis of these results T. H. Morgan obtained in the above crosses he concluded that gene of eye colour trait in Drosophila is present on X-chromosome. The Y chromosome does not carry gene (allele) of eye colour.

### Y-Linked Inheritance or Holandric Traits

Y chromosomes is different from X-chromosome in respect of morphology and genetically. It is not completely inert it was only inert for those traits which are located on X-chromosome. In

Drosophila and human being it carries few genes which have no counterpart on X-chromosome these genes are called Y- linked gene.

### The Y-linked traits are called holandric traits

The alleles of holandric gene are only located at Y-chromosome not on X-chromosome. The Y-linked genes are hemizygous, only located on non-homologous region of Y-chromosome.

They express phenotypically in males, inherit from father to son female do not inherit Y-chromosome so the Y-linked traits cannot pass to them. In human some Y-linked traits porcupine man (Straight hairs on the body), hypertrichosis growth of hairs in the edges of pinna, webbing of toes etc.

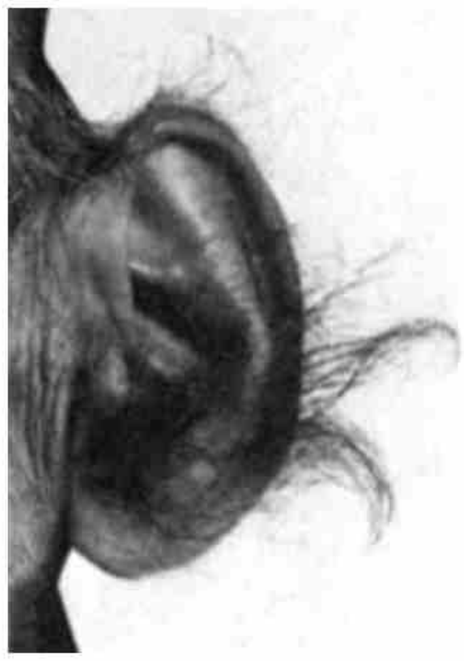


**Fig.23.14 Y-linked trait in human**

### Sex-Limited and Sex-Influenced Traits

We have already studied about sex-linked traits and sex-linked inheritance but some traits are associated with sex of organism i.e. maleness or femaleness there traits are called sex-related traits. This phenomenon of inheritance shows that the genes are not necessary located on sex-chromosomes. These traits may controlled by autosomal or sex-linked genes. There are of sex-related traits i.e. sex-limited and sex-influenced Traits.

### Sex-Limited traits

The type of genes which are present in both sexes of a sexually reproducing organisms but expressed in only one gender (Sex) and turned off in other gender. It is due to anatomical differences in the member of a species e.g. The genes which have horn controlling gene but only expressed in male and female have horn controlling gene but only expressed in male sheep. Another example of sex-limited trait is beard growth in human. It is limited only to men. A women does not grow a beard herself she can pass the genes to her son the purpose of sex-limited gene is to resolve sexual conflict.

## Sex-Influenced Trait

Another type of sex-related traits in that sex-influenced trait, it is an autosomal trait but influenced by sex. These traits are controlled by an allele which is expressed as dominant allele in one but recessive, in other. If a male has one recessive allele it will expressed that character, but in the case of female it requires two recessive to show the same result e.g. **soft facial hairs in female** Vs **coarse facial hairs in male,** another example is baldness which is found in male not in female. It is an autosomal dominant trait in male but autosomal recessive in females, if a male will be heterozygous for this trait he will be bald but a woman should be homozygous for recessive allele to develop baldness.

## Sex-Linkage Inheritance in Man

Human have same pattern of autosomal and sex chromosome. They have XX in female and XY chromosome in male as sex-chromosome. X has many gene on it to control **X-linked traits** like colour blindness, muscular dystrophy hemophilia, congenital deafness, diabetes insipidus etc. some of them are due to recessive alleles and other are due to dominant alleles. Their pattern of inheritance different from each other.

In human genetic studies takes place through mode of inheritance and pedigrees studies because experimental controlled mating is not possible in them.

## X-linked Recessive allelic Inheritance:

The human female which has one X-linked recessive allele of abnormal character called carrier but does not contain any symptoms of this disorder. The male only possess a single X-chromosome if this chromosome carries a recessive allele he will be affected and will be abnormal for that trait. The offspring of carrier female will have 50% chances to receive this abnormal recessive allele, all female children of affected father will be carriers, the male

children of an affected father will not affected at all if their mother is homozygous for normal allele. Example haemophilia and colour blindness.

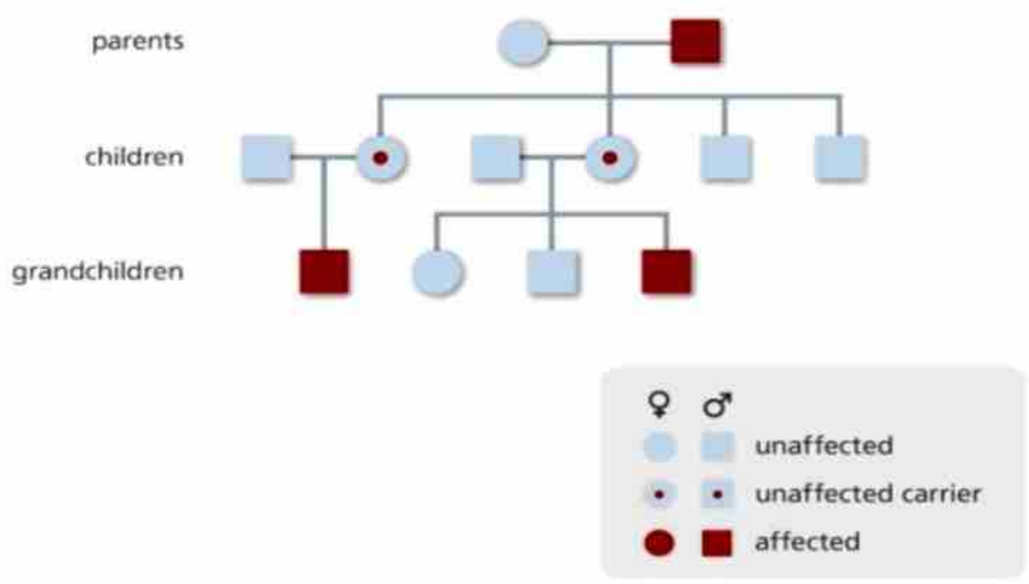
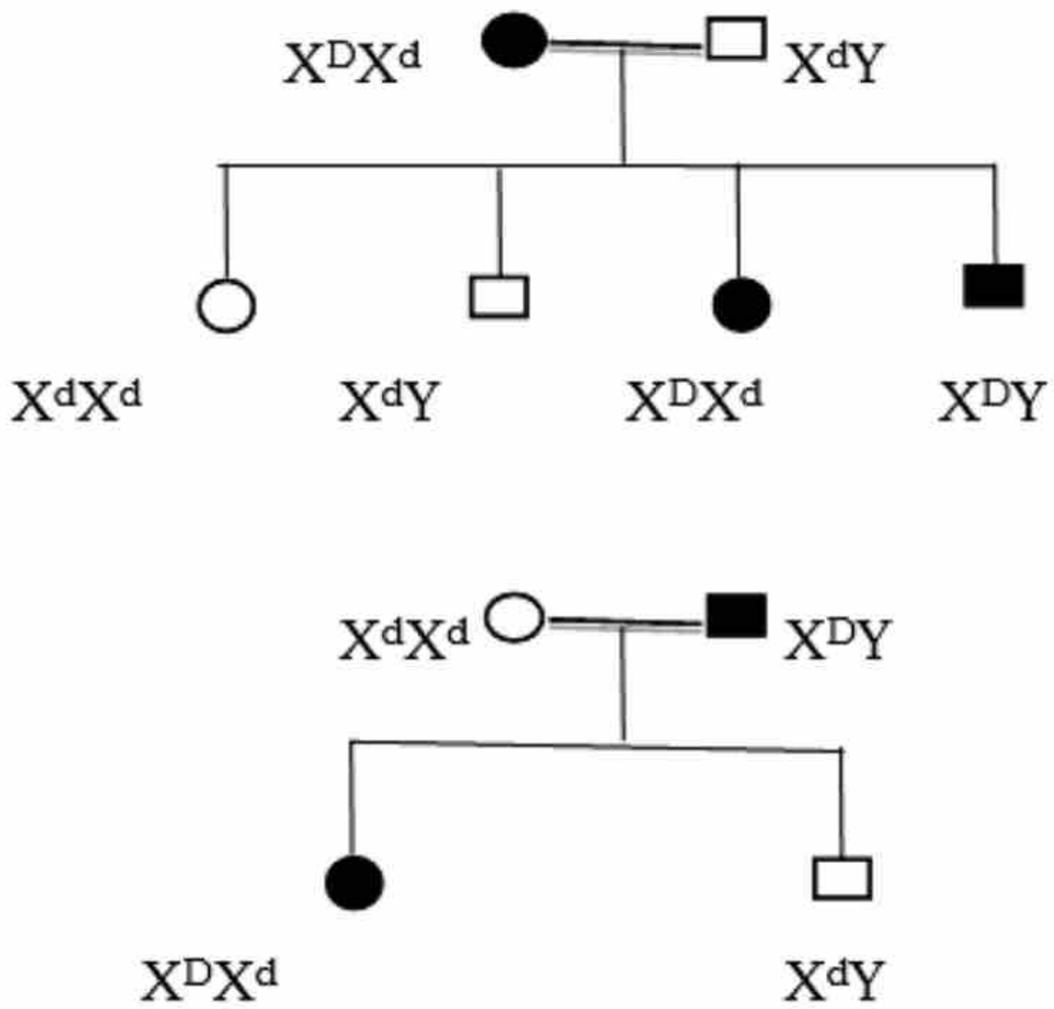



**Fig. 23.15 Pedigree of X-linked recessive trait**

## X-linked Dominant Inheritance

The dominant alleles which are located at X-chromosome are responsible for these types of disorders. In female the affected female may be homozygous or heterozygous. The male and female offspring of a heterozygous affected female has 50% chances of this disorder.

On the other hand if a father has this disorder their all female children will be affected but all male children will be normal e.g. Vitamin D resistant rickets-hypophosphatemia or Rett syndrome



## Genetic Inheritance of Haemophilia

Haemophilia is X-linked recessive human disorder. In these haemophilic persons blood does not clot after injury because the haemophilic persons are unable to synthesize blood clotting factor. There are three types of haemophilia i.e. A, B and C, the A and B are X-linked recessive traits while haemophilia C is an autosomal recessive trait. The chance of X-linked A and B are more in male than female while C affects equally due its autosomal location. The chances of haemophilia is more in male because it is hemizygous in male i.e. control by single allele while in female two doses of alleles are required to develop haemophilia i.e. homozygous recessive. The gene of haemophilia inherits in zig-zag manner from maternal grandfather through a carrier daughter to grandson. It never passes direct from father to son.

Genotype of normal mother = $X^HX^H$

Genotype of haemophilic father = $X^hY$

$P_1$ ♀ Normal female x haemophilic male ♂

$X^HX^H$ $X^hY$

gametes $X^H$ $X^h$ $Y$

$X^HX^h$ $X^HY$

Normal daughter but Carrier of haemophilic gene — Normal son

$P_2$ ♀ Normal female but carrier x Normal male ♂

$X^HX^h$ $X^HY$

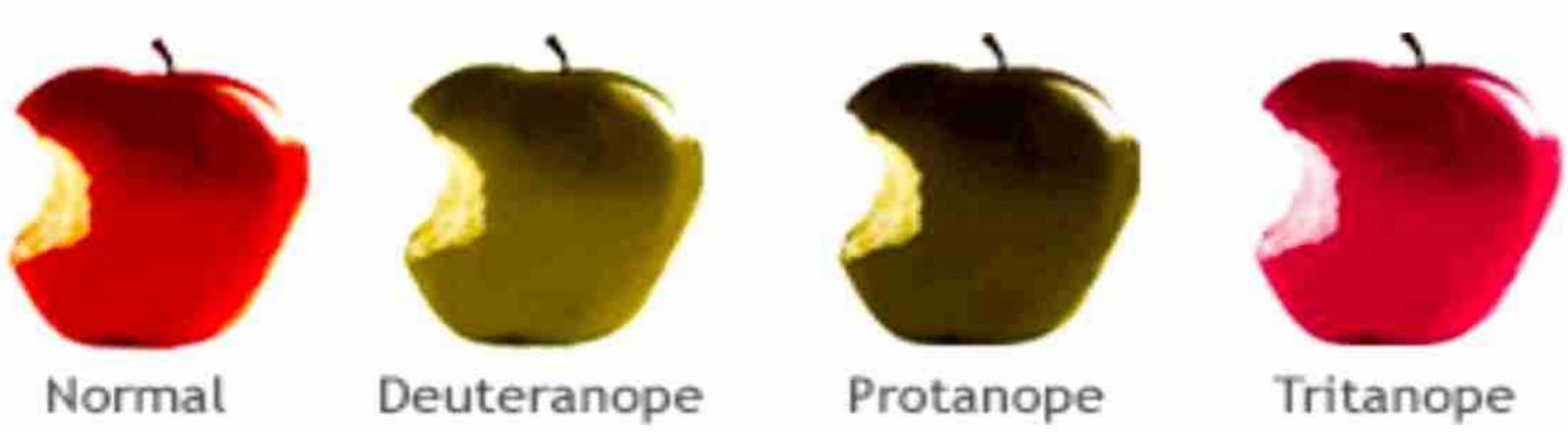
gametes $X^H$ $X^h$ $X^H$ $Y$

| | $X^H$ | $Y$ |
|---|---|---|
| $X^H$ | $X^HX^H$ Normal | $X^HY$ Normal |
| $X^h$ | $X^HX^h$ Normal but carrier | $X^hY$ Haemophilic |

## Inheritance of colour blindness

Human eye has two types of neurons Rod and Cones. Rods are responsible for vision in low light, while cones are responsible colour distinction there are three basic colours normally which develop all colours, the distinct vision of these three colours called to normal trichromatic vision which is due to three different kind of cones located in the retina, each cone in sensitive to only one of these primary-colours i.e. red, green and blue.

Each type of cone has specific light absorbing proteins called **opsins.** These proteins produced by expressing specific genes. The genes for red and green opsins are located on X-chromosomes whereas the gene for blue is located at autosome chromosome no-7. If mutation occurs in these opsins genes develop other recessive alleles for these genes which cause colour blindness in the form of dichromacy and monochromacy.




**Fig,23.16 Dichromacy in human**

**Dichromacy:** It is a condition of colour blindness where a person can distinguish two colours clearly but cannot perceive one of the three, whose gene become mutated and its opsins does not synthesized properly i.e. missing. There are three types of
i) Protanopia is red blindness
ii) Deuteranopia is green blindness
iii) Tritanopia is blue blindness

**Monochromacy:** It is a condition of colour blindness where a person can recognize only one. It is true colour blindness. In blue monochromay only blue colour can be recognized by person but both red and green cone cells opsins are absent. It is X-linked recessive

trait. It is also called red-green colour blindness the inheritance pattern of colour blindness is also like haemophilic pattern of eye colour trait in drosophila. It also work in zig-zag (**criss cross inheritance**) manner and more common in male than female.


**Fig.23.17 Normal and Monochromacy**

## Inheritance of Muscular Dystrophy

It is type of muscle disease caused by mutated allele (gene), the person have this gene have muscle weakness with passage of time which ultimately decrease its mobility with the passage of time. It is sex-linked recessive disorder, the normal gene code for protein called **dystrophin** synthesized in normal person, but missing in the patients of **Duchenne** patient due to recessive alleles, the absence of dystrophin causes leakage of calcium into the cell the symptoms of **Duchene muscular dystrophy** appear in childhood where child begins to have difficulty in standing or rising for standing. Ultimately it become unable to move up to the age of 12 so he or she required wheelchair, with the passage of time they feel difficulty in breathing usually become unable to survive after the age of 20 years, therefore affected male cannot survive for marriage only carrier female survive which can inherit this gene to male baby. This inheritance pattern is similar as haemophilic pattern.

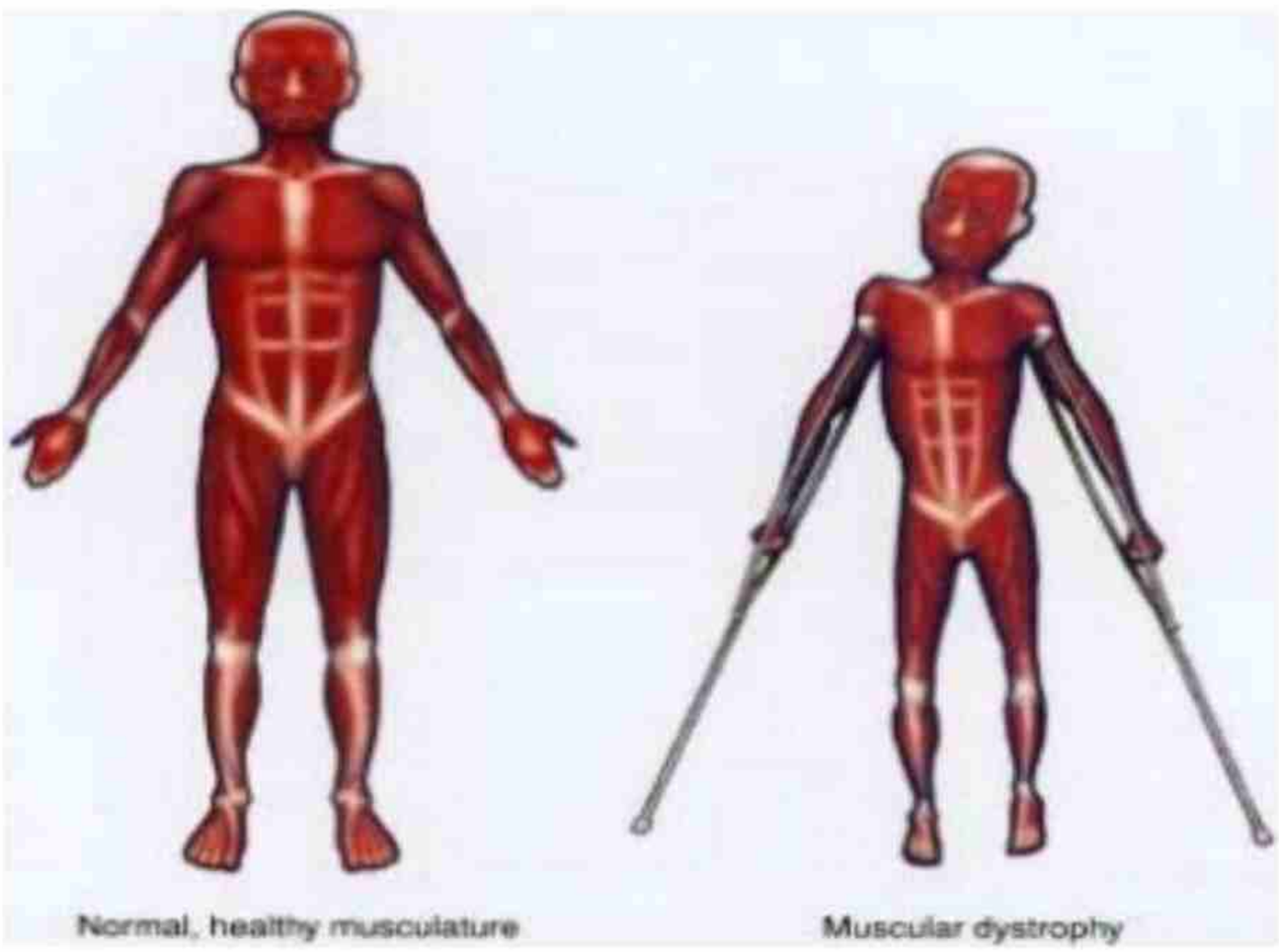



**Fig.23.18 Muscular dystrophy**

## Symbols for pedigree development

- Male
- Female
- Affected Male
- Female
- Carrier Female for sex linked inheritance
- Heterozygous male for autosomal recessive trait.
- Heterozygous female for autosomal recessive trait
- Mating or cross
- Mating or cross
- Dizygotic twins
- Monozygotic twins

## SUMMARY

- The scientific study of inherited characters from parents to offspring, their pattern of inheritance and causes of variation called Genetics.
- The character which was expressed in impure condition (1st generation, F1) called Dominant and its contrasting character, which was not expressed called recessive.
- Cross between phenotypically dominant individual and homozygous recessive to find out the homozygosity or heterozygosity of dominant called test cross.
- Phenomenon of inheritance where expression of both contrasting allele blend in heterozygous condition called incomplete dominance.
- Phenomenon of inheritance where expression of both contrasting allele does not blend heterozygous condition called co-dominance.
- A trait may have more than two alternative forms, the genes which have more than two alternative forms are called multiple alleles.
- International society of blood transfusion has found more than 30 types of blood group system.
- ABO blood group system is controlled by three alleles $I^A$, $I^B$ and i.
- The breakdown of R.B.C produce large amount of bilirubin pigment which accumulate in the foetus and damage the neuron.
- The phenomenon of inheritance where a single trait is controlled by two or more than two separate pairs of genes which manifest themselves in additive manner to yield variable phenotype called polygenic inheritance.
- Labrador is an excellent type of dog its coat colour is one of the best example epistasis in mammals.
- Tendency of gene to remains together in a chromosome called linkage.
- During meiosis the homologous chromosomes pair up, this pairing of homologous chromosome called synapsis.

- Process of exchange of chromosomal segment between non-sister chromatids of homologous chromosome is called crossing over.
- Chromosome which determine sex of the individual called sex-chromosome.
- Chromosomes which are similar in male and female called Autosomes.
- The alleles of holandric gene are only located at Y-chromosome.
- The Y-linked genes are hemizygous, only located on non-homologous region of Y-chromosome.

## EXERCISE

**1. Encircle the correct answer:**

i) Which of the following would cause phenotypic variations among organisms of the same genotype?
(a) Continuous variation within the species
(b) Different varieties of the same species
(c) Different sexes
(d) Exposure to different environments

ii) In which of the following examples of human inheritance is the inheritance pattern explained by multiple alleles on an autosomal chromosome?
(a) The ABO blood group system
(b) Cystic fibrosis
(c) Down's syndrome
(d) Haemophilia

iii) What are the phenotypes of the parents of a colour-blind son and a non-carrier daughter with normal colour vision?

| | Father | Mother |
|---|---|---|
| (a) | Carrier | Normal |
| (b) | Colour-blind | Carrier |
| (c) | Colour-blind | Colour-blind |
| (d) | Normal | Carrier |

iv) In the $F_2$ generation of a dihybrid cross, the phenotypes occurred in the ratio 3:1 what does this result indicate?
(a) The alleles were segregating independently
(b) Polygenic inheritance was involved
(c) Codominance was being shown
(d) The gene loci were linked

v) Two parents, both of blood group A, have a daughter of blood group O. What is the probability that their next child will be a boy who has blood group O?
(a) 0.25 (b) 0.5
(c) 0.75 (d) 1

vi) A boy is colour blind which could be the genotype of his mother?
(a) $X^NX^N$ (b) $X^NX^n$
(c) $X^NY$ (d) $X^nY$

vii) Number of chromosome in grass hopper female is 24. How many chromosomes are present in grass hopper male?
(a) 26 . (b) 25.
(c) 24 (d) 23

viii) The allele of holandric gene is located at?
(a) X-Chromosome (b) Y-Chromosome
(c) Autosome (d) None of these

ix) The gene which interferes and masks the phenotype of the phenotype of the non-locus gene called?
(a) Mutant gene (b) Epistatic gene
(c) Pleiotropy (d) Sex-linked

x) x. International society of blood transfusion has found more than?
(a) 10 blood system (b) 20 blood system
(c) 30 blood system (d) 40 blood system

## 2. Write short answer of the following:

i) Why Rh-incompatibility could be a danger to the developing foetus and mother?
ii) Why haemophilia is common in human male?
iii) What do you mean by sex-influenced trait?
iv) Differentiate between following
   a) Linkage and crossing over
   b) Monohybrid Cross and Dihybrid Cross
   c) X-linked trait and Y-linked trait
   d) Autosomes and sex-chromosome
   e) Incomplete dominance and Co-dominance
v) Why human male are heterogametic?
vi) What do you mean by dichromacy and monochromacy?

vii) What do you known about ZZ and ZW in sex determination?
viii) Why incomplete dominance is called partial dominance?
ix) Define following terms

| | | |
|---|---|---|
| (a) Genetics | (b) Allele | (c) Gene |
| (d) Karyotype | (e) multiple allele | (f) test cross |
| (g) Mutant | (h) Test cross | (i) Back cross |
| (j) Linkage | (k) Crossing over | (l) Protanopi |
| (m) Isoagglutinogen | (n) Erythroblastosis foetilus | |

x) Why law of segregation is called purity of gametes?

## 3. Give detail answer of following questions:

i) Explain epistasis with reference to the inheritance of coat colour in Labrador retriever.
ii) Describe inheritance of two traits with the help of genetic cross.
iii) Describe ABO blood system in human with genetic cross.
iv) Explain sex determination in human and sex linked (x-linked) trait with genetic cross.
v) A male has co-dominant blood group marry with female whose blood group is homozygous A, both have a male child who marry with a female with recessive blood group what will be the chances of blood group in their children? Prove it with genetic cross.