UNIT

22

INHERITANCE

KEY CONCEPTS

The capacity to reproduce is one of the fundamental characteristic of living organisms. It results in the formation of offspring which resembles the parental generation. This tendency of individuals to resemble their parents is known as **heredity**. The resemblance, however is not complete, offspring differ from each other and their parents in many respects. These differences are known as **variations**. Heredity and variations play a significant role in the formation of new species. The science which deals with mechanism of the heredity and variation is called **genetics**. Since the heredity and variation characteristics are controlled by genes, so the term genetics is also referred to the study of genes.

22.1 MENDEL'S LAWS OF INHERITANCE

The science of genetics originated in the year 1900 with the rediscovery of an article originally published in 1866 by an Augustinian monk named Gregor John Mendel. Early philosophers, thinkers and workers had also put forward speculations and theories to explain the mechanism of inheritance but they had not been succeeded in their efforts. Mendel was the first who successfully explained the mechanism of inheritance during his research work on pea plant.

22.1.1 Gregor John Mendel & his work:

Mendel was the pioneer of classical geneticists. Gregor Mendel (1822-1884) was an Austrian monk and is popularly known as the 'Father of genetics'. His experimental work became the basis of modern heredity theory. Mendel was born on July 22, 1822 in Czech Republic. He entered the Augustinian monastery at Brunn, Czech Republic and become actively engaged in investigating variation, heredity and evolution in plants.

Fig: 22.1 Gregor Mendel (1822-1884)

Between 1856 and 1863 he cultivated and tested at least 28,000 pea plants carefully analyzing seven pairs of seed and plant characteristics. He delivered his first lecture on pea experiments in the year 1865 and published his paper 'Experiments on plants hybridization' in the year 1866. His experiments resulted in the formation of the laws of heredity or Mendel's laws. Unfortunately his work made no impression for the next 34 years. Later on in 1900 his work was recognized by three investigators, a Dutch botanist Hugo de Vries, De Correns of Germany and Tschmarck of Austria. He died in Brunn on January 6, 1884.

The reasons why Mendel's work was neglected during his time are as follows:

- The biologists were preoccupied with Darwin's theory of evolution which appeared in the year 1859.

- The journal in which Mendel's work was published was not recognised.
- The scientists of early times were not familiar with the statistical analysis of experimental data.

22.1.2 Mendel's Experiment:

Mendel was very careful about the selection of the plant for his hybridization experiments. He selected the garden pea (*Pisum sativum*) as experimental plant because of the following reasons:

- The plant was easy to grow in pots or in open ground.
- It had a short life cycle.
- The plant has self-pollinating flowers.
- Cross-pollination was also possible.
- The plant possesses distinct contrasting heritable characters.
- Mendel chose seven pairs of characters for his study which is summarized in following table.

Table: 22.1 Seven pairs of contrasting characters in pea plant.

Character	Dominant Trait	Recessive Trait
Seed Shape	Spherical	Wrinkled
Seed colour	Yellow	Green
Flower colour	Purple	White
Pod shape	Inflated	Constricted
Pod colour	Green	Yellow
Flower position	Axial	Terminal
Stem height	Tall	Dwarf

22.1.3 Inheritance of single trait:

One of the reasons of Mendel's success was that he confined to the study of limited traits in pea. In his hybridization (crosses) experiments, first he studied the inheritance of single trait, and then added in his study more traits one by one.

A cross between two individuals that differ with one particular trait is called monohybrid cross. For example, Mendel crossed a true bred round seed shape plant with true bred wrinkled seed shape plant. Such plants are referred as first parental generation (P_1). In first filial generation (F_1) he observed all the offspring with round seed shape.

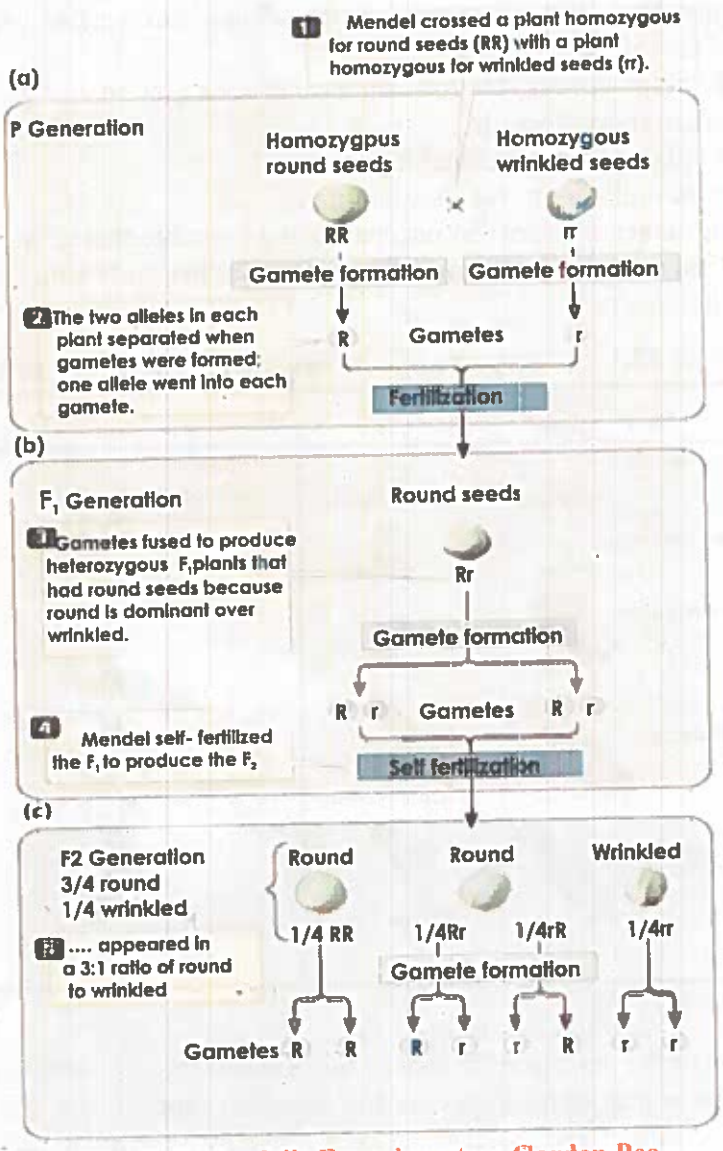


Fig: 22.2 Mendel's Experiment on Garden Pea.

He supposed round seed shape as dominant trait. Next, he allowed the hybrids of F_1 generation to be self-fertilized. In F_2 generation he noticed that both the parental characters appeared. The character that appeared in F_1 was called the dominant character, while hidden character that made its appearance in F_2 was known as the recessive character. The dominant character (in this case round) expressed in approximately 75% offspring whereas recessive (wrinkled in this case) appeared in only 25% of the offspring. He also noticed that the dominant character remained the same even when he conducted reciprocal cross. He tested each of the seven pairs of contrasting characters (given in the table) for the phenomenon of dominance and recessiveness. Mendel observed that the recessive character appeared in the F_2 offsprings in an average ratio of 3:1.

Mendel's predictions:

The science of cytology was in its primitive state during Mendel's time. However, Mendel visualized the cause of inheritance as factors or elements, which were later named as genes by Johannsen in 1909. According to Mendel, each male and female parent contains a pair of factors and each parent passed only one factor of a pair to their offspring. He also predicted that each factor retained its individuality from generation to generation. The factors contributed united randomly to produce the characters of the hybrid. Thus, he indirectly predicted the reduction in chromosome number during gametogenesis and the physical hereditary mechanism.

22.1.4 Mendel's Principles of Inheritance

Mendel did not publish any law as erroneously described in the various books. The rediscoverers of Mendel's work were responsible for our understanding of Mendel's principles. They left that Mendel's work could be represented by laws of heredity. These laws are the law of dominance, law of segregation and the law of independent assortment.

Symbols Used to Represent Mendel's Laws:

According to the classical method of symbolism, the dominant allele is represented by the capital letter while the recessive allele by the small letter. Thus tallness will be represented as 'TT' and recessive character by 'tt'. In the modified method, according to the abnormal recessive allele the symbol is chosen. For example the condition of albinism is characterized by the lack of melanin pigment in the skin, hair, eyes etc. this condition is a rare condition caused by the recessive allele in homozygous condition. The symbol in this case is 'a' for recessive allele and 'A' for the normal allele.

Another method is followed in wild plant bacteria and viruses. When out of two phenotypes, one is more common in the population than its alternative form, it is referred to as the wild phenotype. The rare form is called the mutant phenotype. The symbol "+" is used to indicate the normal allele for wild type and the base letter is borrowed from the name of the mutant type.

Law of Dominance

According to the law of dominance, different characters are controlled by units called factors; factors occur in pairs, of a pair, one factor dominates the other.

Law of Segregation

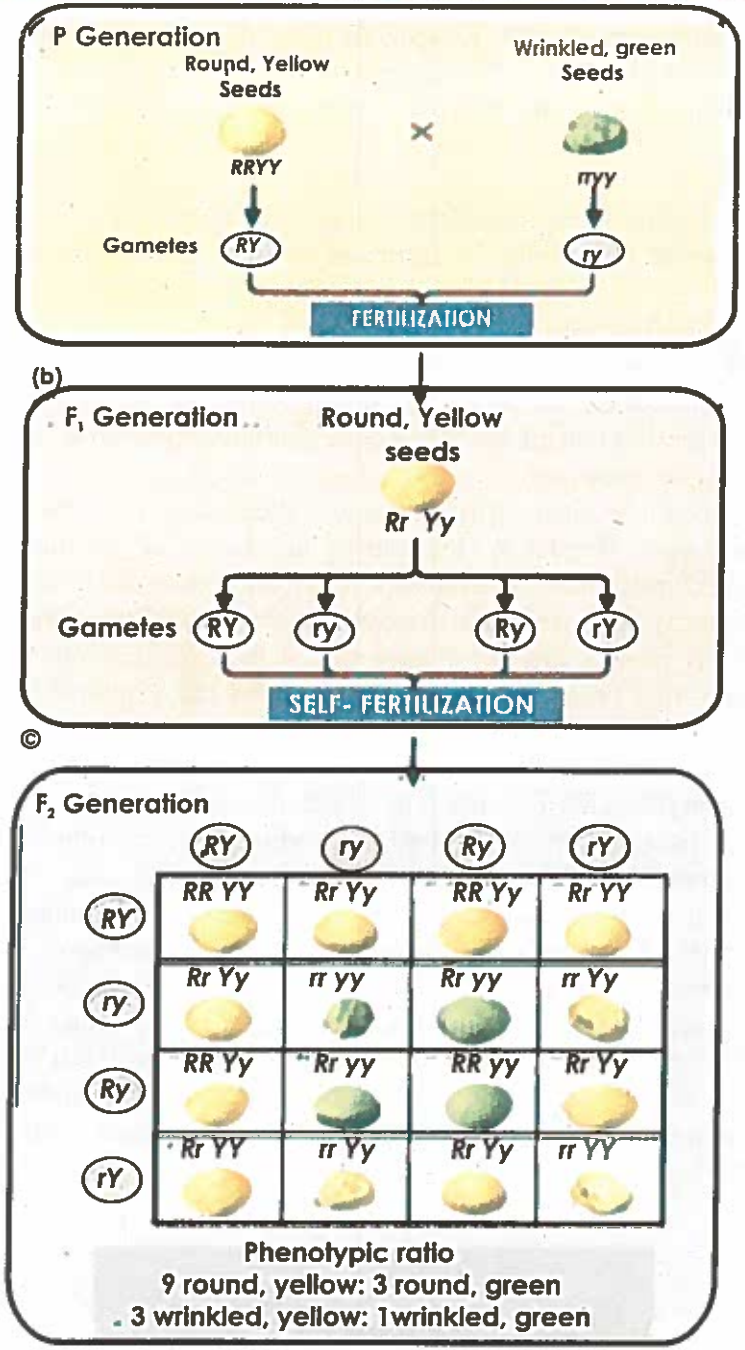
According to the law of segregation or law of purity of gametes, in a heterzygote the dominant and recessive allele remain together without mixing with each other. The alleles separate or segregate from each other during gametogenesis, so that each gamete receives only one allele, either dominant or recessive. During his experiments, Mendel encountered some traits that did not follow the law he had encountered. These traits did not appear independently but always together with at least one other trait. Mendel could not explain what happened and chose not to mention it in his work. Today, it is understood that these traits are because of alleles located on the same chromosome.

22.1.5 Inheritance of two traits:

A dihybrid cross describes a mating experiment between two organisms that are particularly different for two traits. The offspring of such cross is called dihybrid which is heterozygous at two different genetic loci. After understanding the inheritance of single trait, next Mendel decided to study inheritance of two traits simultaneously. For this purpose he performed dihybrid crosses on pea plants and discovered a fundamental law of genetics called the Law of Independent Assortment. Mendel began his experiments by first crossing two homozygous parental organisms that differed with respect to two traits. An organism that is homozygous for a specific trait carries two identical alleles at a particular genetic locus.

Mendel chose to cross a pea plant that was homozygous and dominant for round (RR), yellow (YY) seeds with a pea plant that was homozygous and recessive for wrinkled (rr), green (yy) seeds, represented as RRYY x rryy

The offspring of the RRYY x rryy cross, which is called the F1 generation, were all heterozygous plants with round, yellow seeds and the genotype RrYy. Next, Mendel crossed two plants from the F1 generation. This step is represented as RrYy x RrYy


P Generation
Round, Yellow Seeds
Wrinkled, green Seeds
RRYY
rryy
Gametes
RY
ry
FERTILIZATION
(b)
F1 Generation
Round, Yellow seeds
Rr Yy
Gametes
RY
ry
Ry
rY
SELF- FERTILIZATION
©
F2 Generation
RY
ry
Ry
rY
RR YY
Rr Yy
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Rr yy
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Phenotypic ratio
9 round, yellow: 3 round, green
3 wrinkled, yellow: 1wrinkled, green

Mendel observed that the F2 progeny of his dihybrid cross had a 9:3:3:1 ratio and produced nine plants with round, yellow seeds, three plants with round, green seeds, three plants with wrinkled, yellow seeds and one plant with wrinkled, green seeds.

Conclusion:

From his experiment, Mendel concluded that the pairs of traits in the parental generation assorted independently from one another, from one generation to the next.

22.1.6 Law of Independent Assortment:

The law of Independent Assortment states that two pairs of contrasting traits when followed in a cross, the alleles of one pair assort independently with the alleles of other pair. It implies that alleles of one gene pair have equal probability to with any of the allele of other gene pair.

Independent assortment of genes and their corresponding traits was first observed by Gregor Mendel in 1865 during his studies of genetics in pea plants. Mendel was performing dihybrid crosses, which are crosses between organisms that differ with regard to two traits. He discovered that the combinations of traits in the offspring of his crosses did not always match the combinations of traits in the parental organisms. From his data, he formulated the Principle of Independent Assortment.

We now know that this independent assortment of genes occurs during meiosis in eukaryotes. Meiosis is a type of cell division that reduces the number of chromosomes in a parent cell by half to produce four reproductive cells called gametes. In humans, diploid cells contain 46 chromosomes, with 23 chromosomes inherited from the mother and a second similar set of 23 chromosomes inherited from the father. Pairs of similar chromosomes are called homologous chromosomes. During meiosis, the pairs of homologous chromosome are divided in half to form haploid cells, and this separation, or assortment, of homologous chromosomes is random. This means that all of the maternal chromosomes will not be separated into one cell, while the all paternal chromosomes are separated into another. Instead, after meiosis occurs, each haploid cell contains a mixture of genes from the organism's mother and father.

22.1.7 Scope of independent assortment in variation:

Another feature of independent assortment is recombination. Recombination occurs during meiosis and is a process that breaks and recombines pieces of DNA to produce new combinations of genes. Recombination scrambles pieces of maternal and paternal genes, which ensures that genes assort independently from one another. It is important to note that there is an exception to the law of independent assortment for genes that are located very close to one another on the same chromosome because of genetic linkage

22.1.8 Statistics and Probability Relevant to Genetics

Two basic rules of probability are helpful in solving genetics problems: the rule of multiplication and the rule of addition.

Rule of multiplication (Product rule):

Rule of multiplication is that the probability that independent events will occur simultaneously is the product of their individual probabilities. For example:

Question:

In a Mendelian cross between pea plants that are heterozygous for flower colour (*Pp*), what is the probability that the offspring will be homozygous recessive?

Answer:

- Probability that an egg from the F_1 (*Pp*) will receive a *p* allele = 1/2.
- Probability that a sperm from the F_1 will receive a *p* allele = 1/2.
- The overall probability that two recessive alleles will unite, one from the egg ***and*** one from the sperm, simultaneously, at fertilization is: 1/2 X 1/2 = 1/4.

Rule of addition (Sum rule):

Rule of addition is that the probability of an event that can occur in two or more independent ways is the sum of the separate probabilities of the different ways. For example:

Question:

In a Mendelian cross between pea plants that are heterozygous for flower colour (*Pp*), what is the probability of the offspring being a heterozygote?

Answer:

- There are two ways in which a heterozygote may be produced: the dominant allele (*P*) may be in the egg and the recessive allele (*p*) in the sperm, ***or*** the dominant allele may be in the sperm and the recessive in the egg. Consequently, the probability that the offspring will be heterozygous is the sum of the probabilities of those two possible ways:
- Probability that the dominant allele will be in the egg with the recessive in the sperm is 1/2 X 1/2 = 1/4.
- Probability that the dominant allele will be in the sperm and the recessive in the egg is 1/2 X 1/2 = 1/4.

- Therefore, the probability that a heterozygous offspring will be produced is 1/4 + 1/4 = 1/2.

The Statistical Nature of Inheritance

- If a seed is planted from the F_2 generation of a monohybrid cross, we cannot predict with certainty that the plant will grow to produce white flower (*pp*).
- We can say that there is a 1/4 chance that the plant will have white flowers.
- Stated in statistical terms: among a large sample of F_2 plants, approximately 25% will have white flowers.
- The larger the sample size, the more likely it will be that the percentage of the results will approximate the prediction.

Statistics and Probability

Probability (p) is the chance that an event will occur. Probability equals the case (a) divided by the total number of cases (n) or p = a/n, assuming all cases are equally likely events.

- p for heads? heads/heads plus tails. 1/2
- p for tails? tails/heads and tails. 1/2
- p for heads or tails? 1/2 + 1/2 = 1
- p for heads and tails? These are mutally exclusive. But heads and then tails is possible. 1/2 X 1/2 = 1/4
- **The sum rule:** if ***either/or*** then add the probabilities.
- Die: What is the probability that the die will fall on either 1 or 2 spots? = 1/6 + 1/6.
- **Product rule:** the ***and*** rule, multiply the probabilities.
- What is the probability that die will fall on 1 and next throw, 2? 1/6 X 1/6.

22.2 EXCEPTIONS TO MENDELIAN INHERITANCE

Since Mendel's time, our knowledge of the mechanisms of genetic inheritance has grown immensely. For instance, it is now understood that in how many different ways, alleles interact with their contrasting partner alleles at the same locus. These relationships between the contrasting alleles at the same locus in heterozygous state are called dominance relations. Although Mendel had observed only one form of dominance relation (complete dominance) but later on many geneticists became able to explain several exception to the Mendelian inheritance that could not be explained on the basis of complete dominance. These exceptions are said to have non-Mendelian inheritance patterns. Overall, dominance relations are of following types:

- Complete dominance
- Incomplete dominance

- Co-dominance
- Over dominance

22.2.1 Complete dominance:

When one allele (R) completely dominates the other (r) in heterozygous state is called complete dominance. In this case the heterozygotes (Rr) have the same phenotype as the homozygotes (RR) have. The contrasting pair of alleles for all the seven characters chosen by Mendel showed complete dominance.

22.2.2 Incomplete dominance:

When neither of the two alleles expresses independently in heterozygous state, rather a blend of expression of both alleles is appeared is called incomplete dominance. In 1899 Carl Correns was working on a flowering plant named 4 O'clock. When he crossed a true breeding red flowered plant with a true breeding white flowered 4 O'clock, all the F_1 hybrids had pink flowers. This novel phenotype had a shade intermediate between those of the parents due to an intermediate amount of pigment in petals. When Correns self-fertilized F_1 pink, the F_2 showed all three phenotypes of flower in the ratio of 1 red : 2 pink : 1 white. Red was homozygous for red alleles. and white was homozygous for white alleles. But when allele for red and allele for white were present together in the same plant. neither of them masked the effect of other, rather these alleles showed incomplete dominance in the form of pink colour. As there is no truly dominant allele, the usual capital and small letter distinction for dominant and recessive trait is not necessary. Both the alleles are represented by the same letter "R" but are numbered differently to distinguish white from red.

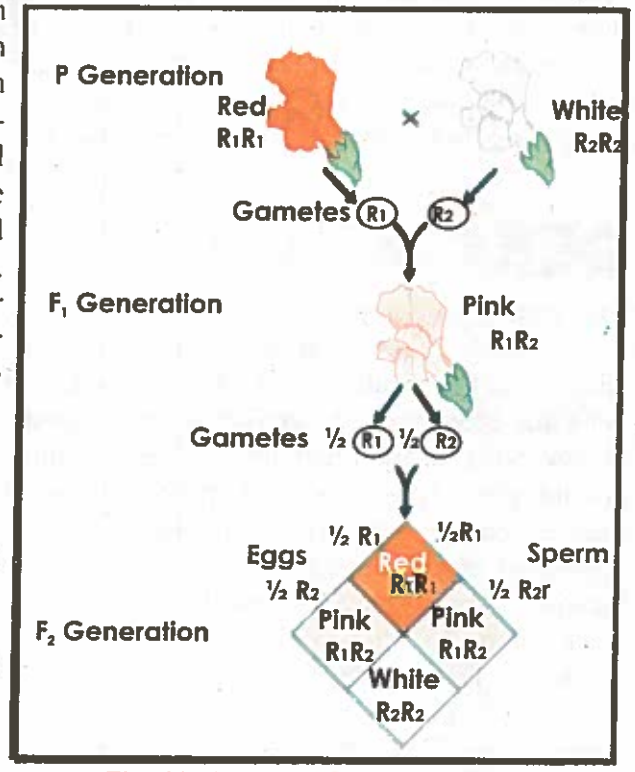


Fig: 22. 4 Incomplete dominance

Allele for red is designated as R_1 and the allele for white as R_2. Punnett square indicates that the phenotypic ratio is the same as the genotypic ratio.

There is absolutely no need of test cross.

22.2.3 Co-dominance:

It is not an intermediate quantitative expression like incomplete dominance but in this case different alleles of a gene that are both expressed in a heterozygote condition are called co-dominant. For example, in case of blood group AB both antigen A and antigen B are expressed and the group is named as blood group AB.

22.2.4 Over dominance:

When the phenotypic expression of heterozygotes (w^+/w) become more intense than the homozygous (w^+/w^+) state of the dominant allele, is called over dominance. For example, in fruit fly (*Drosophila*) the heterozygote (w^+/w) has more quantity of florescent pigments in eyes than wild (w^+/w^+) or white eye (w/w) homozygote.

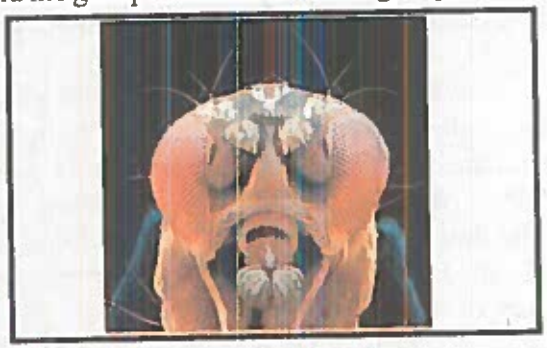


Fig: 22. 5 Coloured scanning electron micrograph (SEM) of the head of a fruit fly (Drosophila sp.). Its two compound eyes (red) are seen on either side of its head.

22.3 ABO BLOOD GROUP SYSTEM

22.3.1 Multiple Alleles:

Allele is the form of a gene which codes for one possible outcome of a phenotype. For example, in Mendel's pea investigations, he found that there was a gene that determined the colour of the pea pod. One form of it (one allele) creates yellow pods, & the other form (allele) creates green pods. It indicates that two possible phenotypes of one trait (pod colour) are determined by two alleles (forms) of the one "colour" gene. If a trait exists in three different phenotypes, there must be some sort of interactions like incomplete dominance or co-dominance is found between the two alleles. Now, if there are 4 or more possible phenotypes for a particular trait, then more than 2 alleles for that trait must exist in the population. All of such alleles are called "multiple alleles".

Multiple alleles are produced by gene mutation. Some genes may have as many as 300 alleles, but individuals have only two of those alleles. Why? Because individuals have only two biological parents. We inherit half of our genes (alleles) from mother, & the other half from father, so we end up with two alleles for every trait in our phenotype.

An excellent example of multiple allele inheritance is human ABO blood group system. It is the most well-known blood group system which was discovered in 1901 at the University of Vienna by Karl Landsteiner in the process of trying to learn why blood transfusions sometimes cause death and at other times save a patient. In 1930, he received the Nobel Prize for his discovery of blood types.

22.3.2 ABO blood groups:

ABO blood groups are found in all humans and in many other primates such as apes chimpanzees, baboons, and gorillas. There are four principal types: A, B, AB, and O, there are two antigens and two antibodies that are mostly responsible for the ABO types. The specific combination of these four components determines an individual's type in most cases.

Table: 22.2 ABO blood groups

ABO Blood type	Antigen (A or B)	Antibodies (Anti-A or Anti-B)	Donors
A	A	Anti-B	A,O
B	B	Anti-A	B, O
AB	Both A&B	None	A,B,AB,O
O	None	Both Anti-A or Anti-B	Only O

For example, people with type A blood will have the A antigen on the surface of their red blood cells (as shown in the table below). As a result, anti-A antibodies will not be produced by them because they would cause the destruction of their own blood.

However, if B type blood is injected into their systems, anti-B antibodies in their plasma will recognize it as alien and burst or agglutinate the introduced red cells in order to cleanse the blood of alien protein.Individuals with type O blood do not produce AB antigens. Therefore, their blood normally will not be rejected when it is given to others with different ABO types. As a result, type O people are universal donors for transfusions, but they can receive only type O blood themselves.

Those who have type AB blood do not make any antibodies. Their blood does not discriminate against any other ABO type. Consequently, they are universal recipients for transfusions, but their blood will be agglutinated when given to people with every other type because they produce both kinds of antigens.

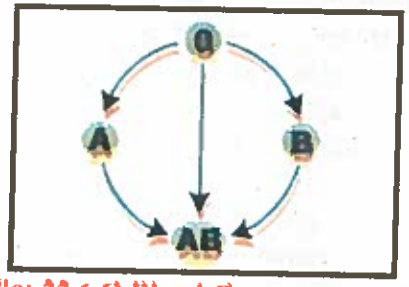


Fig: 22.6 O Blood Group people are universal donors for transfusions

22.3.3 Genetic basis of ABO blood groups:

Bernstein explained the genetic basis of ABO system in 1925. It is a multiple allelic trait that is encoded by a single polymorphic gene "I" on chromosome 9. This gene exists in three multiple alleles that have been produced through mutation.

Gene "I" → Mutation 1 → I^A (antigen-A producing allele)
Gene "I" → Mutation 2 → I^B (antigen-B producing allele)
Gene "I" → Mutation 3 → i (non-antigen producing allele)

Table: 22.3 Genetic basis of ABO blood groups

ABO Blood Type	Genotypes
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B
O	ii

Their dominance relations are very interesting, alleles I^A and I^B are completely dominant over the allele i, while I^A and I^B are co-dominant to each other because each expresses in equally in heterozygous (I^AI^B) state to produce AB phenotype. Therefore I^AI^A or I^Ai genotypes will produce phenotype A. Similarly I^BI^B or I^Bi produces phenotype B. The homozygous ii will produce phenotype O.

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

22.3.4 Occurrence of some other blood group systems:

There are a number of different blood group systems found in human. The International Society of Blood Transfusion has recognized up to 30 major group systems. These systems are characterized by the presence or absence of specific molecules, called antigens that are situated on the surface of the red blood cells. Most antigens are protein molecules. Two main blood group systems are ABO system and Rh (Rhesus) system. These two systems are more significant because incompatibility between donor and recipient's blood with respect to these two systems may become dangerous to life.

In the "ABO" system, all blood belongs to one of four major groups: A, B, AB, or O; and in "Rh" system, there are two groups i.e. Rh positive or Rh negative. But there are more than two hundred minor blood groups (belong to the rest of blood group system other than ABO & Rh systems) that usually do not complicate the blood transfusions. These are known as rare blood types.

Table: 22.3 Physiology of Rh blood group.

Rh Blood Type	Rh-factor or antigen	Anti-Rh Antibodies	Donors	Genotypes
Rh^+	Present	Absent	Rh^+/Rh^-	DD/ Dd
Rh^-	Absent	Present	Only Rh	dd

Examples of blood group systems other than "ABO" and "Rh" are MNS, P, Lutheran, Kell, Lewis, Duffy, Kidd, Diego, etc.

22.4 Rh BLOOD GROUP SYSTEMS

The Rh (Rhesus) blood group system (including the Rh factor) is one of the currently 30 human blood group systems. It is clinically the most important blood group system after ABO. The name of this system (Rh) is derived from *Rhesus* monkey, because its antigen was first discovered in it by Landsteiner in 1930s.The Rh blood group system currently consists of 50 defined blood-group antigens, among which the 5 antigens D, C, c, E, and e are the most important ones.

The commonly-used terms Rh factor, Rh positive and Rh negative refer to the D antigen only. So the persons having this antigen are called Rh positive and those in which it is absent are called Rh negative. Other antigens of Rh blood group system have no significant role in blood transfusion.

On the other hand the D antigen incompatibility between donor and recipient can cause problem not only during blood transfusion but it is also a relevant cause of the hemolytic disease of the newborn or erythroblastosis foetalis.

For Your Information

ABO Blood type antigens are not only found on the surface of red cells. They are also normally secreted by some people in their body fluids, including saliva, tears, and urine. Such persons are called secretors. Whether someone is able to secrete them is genetically controlled by a dominant secretor gene "Se" presents on chromosome 19.

22.4.2 Genetic basis of Rh blood group system:

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 genes *C* and *E* alternatively occupy the other locus. The D locus is of prime importance, because it is associated with the formation of D antigen (commonly known as Rh factor).

Gene *D* has two alleles, *D* and *d*. *D* is completely dominant over *d*. persons having genotypes *"DD"* or *"Dd"* have D antigen (Rh factor) on their RBC and are Rh positive. Persons with genotype *"dd"* don not have Rh factor and are Rh negative.

22.4.3 Maternal-foetal Rh incompatibility:

The Rh factor is notorious in cases where antibodies produced by a pregnant woman react with the red blood cells of her developing foetus. The situation arises if the mother is Rh-negative but the foetus is Rh-positive, having inherited the factor from the father. The mother develops antibodies against the Rh factor when small amounts of fetal blood cross the placenta and come in contact with her lymphocytes, usually late in pregnancy or during delivery of the baby. Typically, the mother's response to this first exposure is mild and without medical consequences for the baby. The real danger occurs in subsequent pregnancies, when the mother's immune response against the Rh factor has already been exposed and her antibodies can cross the placenta and destroy the red blood cells of an Rh-positive foetus. This condition is referred as erythroblastosis foetalis. To prevent this, the mother may be injected with anti- Rh antibodies after delivering her first Rh-positive baby.

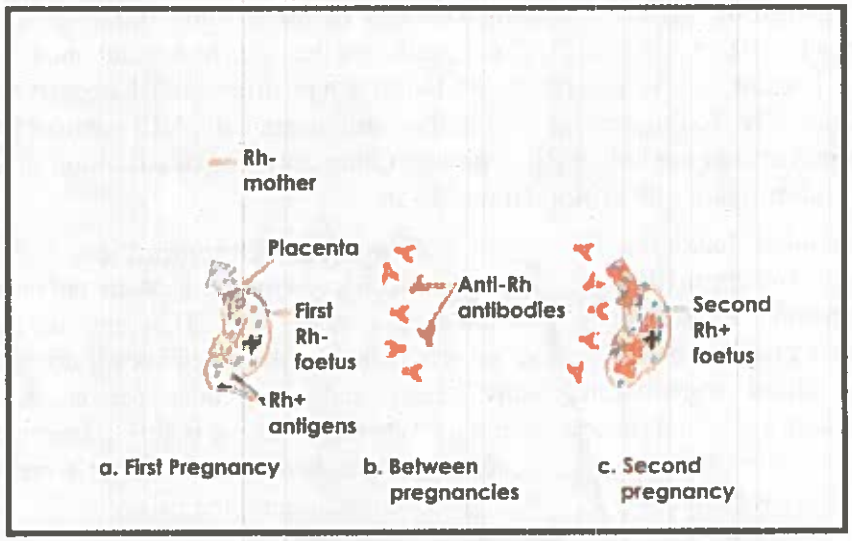


Fig: 22.6 Erythroblastosis foetalis

The antibodies destroy the Rh-positive red cells that have entered the mother's circulation before her own immune system has been stimulated by the Rh antigen.

DO YOU KNOW?

An **erythroblast** is a type of red blood cell which still retains a cell nucleus. It is the immediate precursor of a normal erythrocyte.

22.5 GENE INTERACTIONS

During the study of Mendelian experiments, you have learnt about the phenomenon of dominance which is an interaction between the contrasting alleles of same locus. Now you are going to understand the interaction between the alleles of different gene pairs located on different loci of same or different chromosomes. These are known as **non- allelic interactions** or **inter-genic interactions**.

22.5.1 Epistasis:

An example of non- allelic interactions is **epistasis**, which can be defined as the phenomenon in which the effect caused by the genes at one locus interferes with or hides the effect caused by another gene at another locus. In such interactions, the gene which suppresses or masks the effect of action of a gene at another locus is known as **epistatic gene** and the gene which is suppressed is known as **hypostatic gene**.

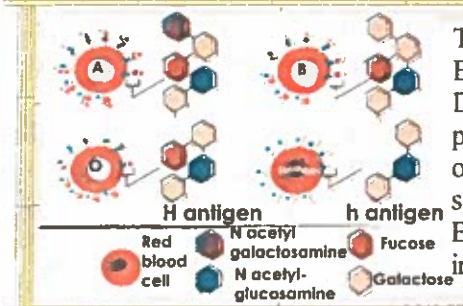
Epistasis must not be confused with dominance. Dominance is the relationship between alleles of the same gene occupying the same locus, but epistasis is the interaction between different genes occupying different loci. Following table further illustrate the difference between epistasis and dominance.

22.5.2 Bombay phenotype:

The Bombay phenotype in humans is an example of epistasis. This is an altered and very rare blood group phenotype in which individuals are phenotypically O but genotypically they may be like A, B, or AB. Actually the production of A or B antigen also dependent upon H-substance, which is synthesize by another gene *"H"* on chromosome 19. Its recessive allele *"h"* does not synthesize this substance.

DO YOU KNOW?

This blood phenotype was first discovered in Bombay, now known as Mumbai, in India, by Dr. Y.M. Bhende, as published in 1952. It is present in about 0.0004% (about 4 per million) of the human population generally, though in some places such as Mumbai (formerly Bombay) local populations can have occurrences in as much as 0.01% (1 in 10,000) of inhabitants.



Bombay

The H-substance is a precursor to the A and B antigens. For instance, the *B* allele must be present to produce the B enzyme that modifies the H-substance to become the B antigen. It is the same for the *A* allele. However, if only recessive alleles for the H-substance are inherited *(hh)*, as in the case of Bombay phenotype,

the H-substance will not be produced. Subsequently, the A and B antigens also will not be produced.The result is an O phenotype by default since a lack of A and B antigens is the O type. This means that the ABO genotypes also require *HH* or *Hh* genotypes for their correct expression.

So the individuals who are homozygous recessive at the H-locus *(hh)* will be blood type O regardless of the genotype at the *I*-gene where the alleles for ABO blood types reside.

ACTIVITY

Consider a child from a type AB male and a type O female who are both heterozygous carriers of the recessive h-allele. What is the probability that this couple will have a type O child?

22.5.3 Polygenic Inheritance:

Genotypes interact with environment to produce phenotypes. Phenotypic expressions of the traits are of two different types i.e. qualitative and quantitative. **Qualitative traits** have few phenotypes that have sharp and more obvious differences among them so they show discontinuous variations.

On the other hand **quantitative traits** comparatively have large number of phenotypes that have small and less striking difference so they have continuous variations.

For instance, some traits like pea seed shape exists in two sharply distinct phenotype, round and wrinkled; others like 4 O'clock flower colour can have three phenotypes, red, pink and white; still others like ABO blood group system have four qualitatively different phenotypes, A, B, AB, and O.

But there are number of traits such as height, weight, intelligence, and skin colour in human; and grain colour in wheat exhibit continuous quantitative variations over the wide range of many phenotypes from one extreme to another.

Mendel focused on traits that showed only two qualitatively different phenotypes which could be determined by just two alternate alleles of a single gene. But a wide range of phenotypes of a quantitative trait cannot be encoded by a single gene with two alleles.

Even a few multiple alleles of a single gene cannot make a large number of phenotype. In fact, 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**. These quantitative traits are therefore called **polygenic traits.**

All the genes that control a quantitative trait are called **polygenes** which have a small positive or negative effect on the character. Polygenes supplement each other and sum of positive and negative effect of all individual polygenes produce quantitative phenotype of a continuous varying traits.

22.5.4 Wheat grain colour:

Wheat grain colour is a good example of polygenic (multiple gene) inheritance. Wheat grains show a continuous variation in colour from white to dark red.

Approximately seven different phenotypes are found in wheat population all over the world. Some grains are white, some are deep red but most grains have shades in between from light pink to moderately dark red. Nilsson Elle 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 F_1 grains had light red colour intermediate between two parental shades.

It seemed as if it was a case of incomplete dominance. But 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 following ratio:

Dark red	Moderately dark red	Red	Light red	Pink	Light Pink	White
1	6	15	20	15	6	1

Three different gene pairs, 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 a negative effect.

If all the six allele code for red pigment (AABBCC), the grain is dark red; when none of the six allele encodes red pigment (aabbcc), the grain is white. When a grain has one allele for red pigment (Aabbcc or aaBbcc or aabbCc) its colour is light pink; if it has two alleles for the pigment (AaBbcc or aaBbCc or AabbCc) it is pink, if it has three pigment alleles (AaBbCc or AABbcc or AabbCC), it will be light red. Similarly four alleles colour dose (AABBcc or aaBBCC or AAbbCC) will make red and five alleles colour dose (AABBCc or AABbCC or AaBBCC) will produce moderately dark red grain.

Thus colour phenotypes of grains depend upon the number of pigment producing alleles (A, B, and C). Environmental factors like light, water and nutrients also influence the amount of grain colour.

22.5.5 Human skin colour:

Human skin colour is a good example of polygenic (multiple gene) inheritance. Skin colour is largely determined by the amount of melanin the skin produces. Dark-skinned individuals produce more melanin than light-skinned individuals. At least three genes regulate the amount of melanin produced.

- Gene A is involved in the permanent survival, proliferation and migration of melanocytes.
- Gene B encodes the enzyme tyrosinase which is involved in the production of melanin from tyrosine.

- Gene C is primarily responsible for determining whether pheomelanin or eumelanin is produced in humans.

Each gene has two forms: dark-skin allele represented by capital case letters (A, B, and C) and light-skin allele represented by small case letters (a, b, and c). No allele is completely dominant to the other, and heterozygotes exhibit an intermediate phenotype (incomplete dominance).

A, B, and C act as dark-skin alleles in the genotype, because they add pigment by increasing melanin production. On the other hand a, b, and c act as light-skin alleles in the genotype because they inhibit melanin production.

There are seven different shades of skin colour ranging from very light (aabbcc) to very dark (AABBCC); most individuals have the intermediate skin colour (AaBbCc). This latter genotype would be characteristic of a mulatto (an offspring of a black and a white parent). In the cross between two mulatto genotypes (AaBbCc X AaBbCc), each parent produces eight different types of gametes and these gametes combine with each other in 64 different ways resulting in a total of seven skin colours. The skin colours can be represented by the number of capital letters, ranging from zero (no dark skin alleles) to six (all dark skin allele). The approximate shades of skin colour corresponding to each genotype are shown in Table 22.4

Table: 22.4 Different shades of skin colour.

Pigment alleles colour shade	6	5	4	3	2	1	0
	Dark Brown	Moderately dark brown	Brown	Light Brown	Pinkish Brown	Whitish brown	Pure white
Ratio	1	6	15	20	15	6	1

22.6 GENE LINKAGE AND CROSSING OVER

22.6.1 Gene linkage:

The number of genes in a cell is far greater than the number of chromosomes; in fact each chromosome has hundreds and thousands of genes. Genes located on the same chromosome that tend to be inherited together in genetic crosses are said to be **linked genes**, and the phenomenon of staying together of more than one gene on the same chromosome is called **gene linkage**. If genes are linked on autosomes, their linkage is called autosomal linkage. Similarly, if they are linked on sex chromosome, their linkage is called sex linkage.

All the linked genes found on the same homologous pair of chromosome form a group, known as **linkage group**, so the number of linkage groups in an organism are equal to the number of homologous pair of chromosomes in that organism.

For instance, in human, the genes for sickle cell anaemia, leukemia, and albinism are found on chromosome 11, so these traits are supposed to be in the same linkage group.

As it is mentioned earlier, that linked genes tend to be inherited together (*en bloc* inheritance) in the offspring, so usually they do not show recombination and do not assort independently in the offspring. So the ideal Mendelian ratio of independent assortment is deviated.

22.6.2 Detection of gene linkage:

Gene linkage can easily be detected by performing a test cross between two gene pairs (dihybrid test cross). In such type of test cross, a heterozygous individual for two traits (F_1) is back crossed with its recessive parent (P_1). If all four phenotypic combinations (parental & recombinants) are produced in equal 1:1:1:1 ratio, then there would be no linkage between the genes. If this ratio is deviated i.e. more parental types and less recombinant types, this indicates the **incomplete or partial linkage**; but if only parental types are produced, **complete or tight linkage** is believed.

To see how linkage between genes affects the inheritance of two different characters, let's take an example from T. H. Morgan's experiments on Drosophila.In Drosophila, the shape of wings exists in two forms; normal wing shape is dominant over vestigial wing.

Similarly, body colour also exists in two forms; the grey body colour is dominant over black body colour. When Morgan made a cross between the individual having grey body and normal wings with another individual having black body and vestigial wings, all the F_1 progeny inherited grey body and normal wing phenotypes. When these flies were test crossed with their P_1 recessive, following results were observed:

- Grey body and normal wings (parental type) = 965
- Black body and vestigial wings (parental types) = 944
- Grey body and vestigial wings (recombinant types) = 206
- Black body and normal wings (recombinant types) = 185

Since most offspring had a parental phenotype (incomplete / partial linkage), Morgan concluded that the genes for body colour and wing size are located on the same chromosome. However, the production of a relatively small number of offspring with nonparental phenotypes indicated that some mechanism occasionally breaks the linkage between the genes on same chromosome.

Later on it was discovered that crossing over at the time of meiosis (gamete formation in animals and spore formation in plants) is the mechanism that occasionally breaks the linkage between the genes on same chromosome.

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22.6.3 Crossing over:

Subsequent experiments demonstrated that the process which is responsible for the recombination of linked genes is crossing over. In crossing over, an exchange of maternal and paternal chromatid parts occurs while homologous chromosomes are paired during prophase of meiosis I. The recombinant chromatids resulting from crossing over may bring alleles together in new combinations, so when they are distributed in different gametes, a wide variety of gametes are produced.

Let's recall Morgan's experiment in which a female fly having grey body and normal wings was crossed by a male fly having black body and vestigial wings. Although most of the eggs had a chromosome with either the $b^+ vg^+$ or b vg parental genotypes for body colour and wing size, but some eggs had a recombinant chromosome with $b^+ vg$ or $b\ vg^+$ genotypes. Fertilization of these various classes of eggs by homozygous recessive sperms (*b vg*) produced an offspring population in which 17% exhibited nonparental, recombinant phenotypes. These recombinants were the products of crossing over.

22.6.4 Recombination frequency & Genetic Map of chromosome:

Discovery of linked genes and recombination due to crossing over led one of Morgan's students, Alfred H. Sturtevant, to a method of constructing a **genetic map** or **linkage map**, an ordered list of genetic loci along a particular chromosome.

Sturtevant hypothesized that recombination frequencies, which are the result of crossing over, depend upon the distance between the linked genes on chromosomes. So he assumed that the farther apart two genes are, the higher the probability that a crossover will occur between them and therefore the higher the recombination frequency. The recombination frequency is determined as follows:

$$\text{Recombination Frequency \%} = \frac{\text{Sum of recombinants}}{\text{Sum of all combination (Parental + Recombinants)}} \times 100$$

Using recombination data from various fruit fly crosses, Sturtevant assign relative positions of several genes along the length of chromosome. For instance, figure 22.8 shows the Sturtevant's linkage map of three genes: the body colour *(b)*, wing size *(vg)*, and a third gene, called cinnabar *(cn)*. Cinnabar is one of many Drosophila genes affecting eye colour. Cinnabar eyes, a mutant phenotype, are a brighter red than the wild type colour. The recombination frequency data is given below:

- $cn-b = 9\%$
- $cn-vg = 9.5\%$
- $b-vg = 17\%$

With the help of these data, the only assumption about the location of these three genes can be made is that *cn* is located midway to the *b* and *vg*. Sturtevant expressed the distances between genes in **map units.**

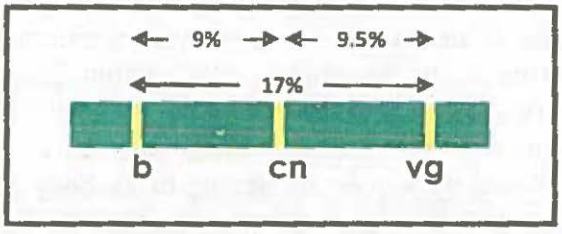


Fig:22.8

The map units are arbitrary, so they are not related to any physical units of length, however, one map unit is supposed to equal to 1% recombination frequency. Today map units are often called ***centimorgans (cM)*** in honor of Morgan.

The b-vg recombination frequency (17%) is slightly less than the sum of the b-cn and cn-vg frequencies (9 + 9.5 = 18.5%) because of the few times that crossover occurs between b and cn and an additional crossover occurs between cn and vg. The second crossover would "cancel out" the first, reducing the observed b-vg recombination frequency while contributing to the frequency between each of the closer pair of gene. The value of 18.5% (18.5 map units) is closer to the actual distance between the genes, so a geneticist would add the smaller distances in constructing a map.

22.7 SEX DETERMINATION

Whether we are male or female is one of our more obvious phenotypic characters. Although the anatomical and physical differences between women and men are numerous, the chromosomal basis for determining sex is rather simple. The search for mechanism of inheritance of sex started after discovery of Mendel's work in 1900. A clear picture of the genetic basis of sex determination emerged after the discovery of sex chromosome.

22.7.1 Patterns of Sex Determination:

There is a wide variety of sex determining mechanisms but three patterns are more common.

XO – XX Type:

This pattern of sex determination is found in grasshopper and Protenor bug. 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 and the other half without any sex chromosome. A gamete without any sex chromosome is called nullo gamete.

Female is XX, because it has two X chromosomes. It is homogametic, as it forms only one type of eggs. Every egg carries an X chromosome. Sex of the offspring depends on the kind of sperm that fertilizes the egg. If an X-carring 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.

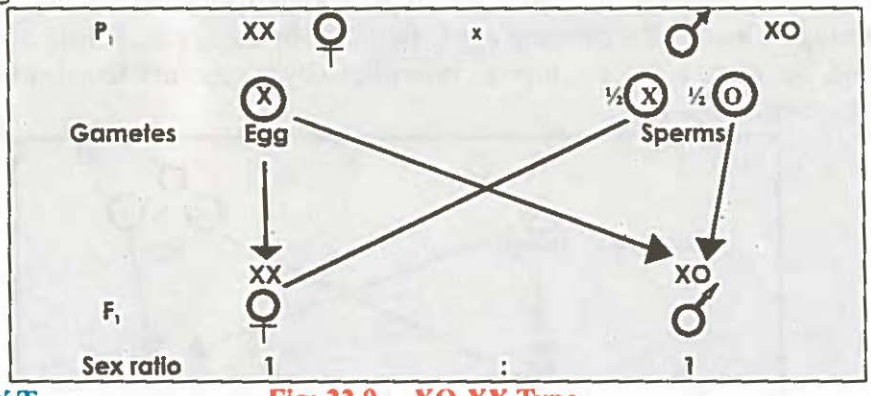


Fig: 22.9 XO-XX Type

XY – XX Type:

This pattern of sex determination is found in Drosophila, human and many other organisms. Male is XY and female is XX. Male being heterogametic produces two types of sex-determining sperms. Half the sperms carry X-chromosome and the other half carry Y – chromosome. Chances for both types of sperms are equal.

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 XX, and a female offspring is produced. If a Y – 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 1:1. Sex ratio indicates chances of the sex of the offspring. Chances for a son or daughter in human birth are equal.

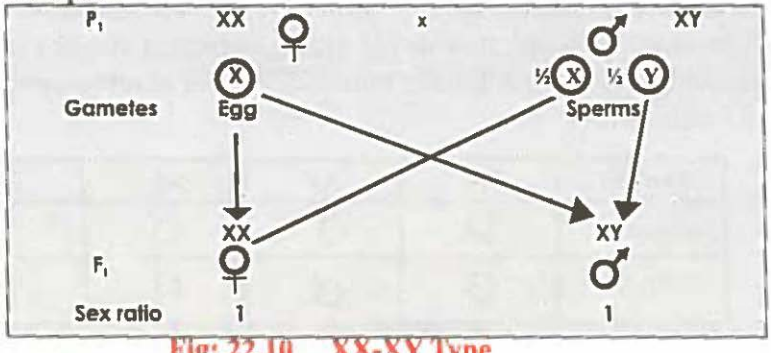


Fig: 22.10 XX-XY Type

XX – XY type or WZ – ZZ Type:

This type of sex – determination pattern is common in birds, butterflies and moths. It was discovered by J. Seiler in 1914 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 is 1:1

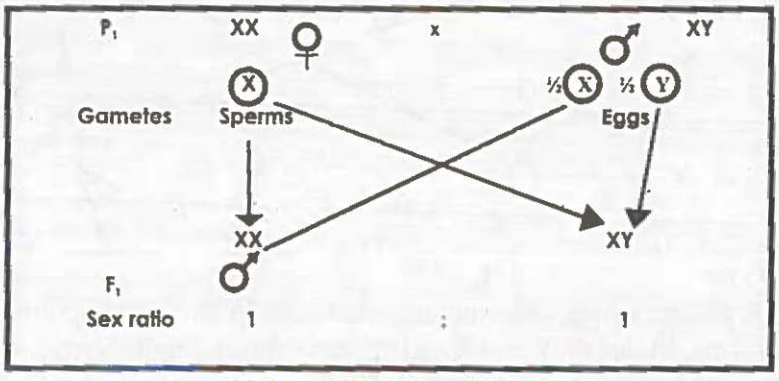


Fig: 22.11 XX-XY or WZ-ZZ Type

22.7.2 Comparison of chromosomal determination of sex between Drosophila and Humans:

Although both *Drosophila* and humans follow the same XY – XY sex determining pattern, yet there is a basic technical difference between the two. Presence of 'SRY' gene on Y chromosome is essential for triggering the development of maleness in humans. Absence of Y chromosome simply leads to the female development path. XO Turner's syndrome in humans produced through non-disjunction is a sterile female. But in *Drosophila* XO is a sterile male. Similarly XXY individual produced through non disjunction gametes in humans is a sterile male called Klinefelter's syndrome, but the same XXY set of chromosomes in Drosophila produces a fertile female.

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

There is a close genic balance between genes of different chromosomes. *Drosophila* has an X chromosome-autosome balance system. Its Y chromosome appears to have little influence on sex.

Here actually the X chromosome is female determining and the autosomes are male determining. 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.

22.8 SEX LINKAGE

22.8.1 Sex Linkage in Drosophila

T. H. Morgan made the first demonstration of a sex-linked trait in 1910. Through his experiments on *Drosophila*, the common fruit fly, Morgan showed that the inheritance of eye colour and sex occurs in a coordinate fashion. He reasoned correctly that the eye colour gene is located on the X chromosome but is not present on the Y chromosome. This meant that the recessive allele specifying white eyes is always expressed in males but could be masked by the presence of a dominant red eye colour allele in heterozygous females.*Drosophila melanogaster* eye colour turned out to be a most informative trait. At first, all the flies Morgan raised were wild type for eye colour; they had brick –red eye (wild type).

In 1910, a white-eyed male appeared in a laboratory bottle. Apparently the variant form arose through a spontaneous mutation in a gene controlling eye colour. Morgan established true breeding traits of white-eyed males and females. Then he did a series of reciprocal crosses represented in the following figures.

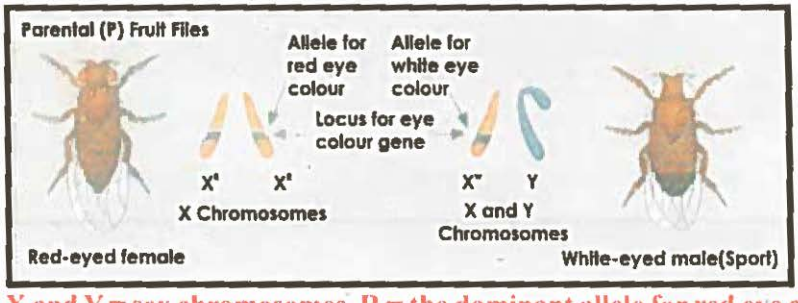


Fig 22.12 X and Y = sex chromosomes, R = the dominant allele for red eye colour, w = the recessive allele for white-eye colour; X^R indicates that the X chromosomes carries the allele for red eyes; X^w indicates that the X chromosomes carries alleles for white eyes. The Y-chromosomes has no locus for eye colour genes. These are the phenotypes, sex chromosomes and alleles for eye colour in Morgan's experimental fruit flies in the P generation.

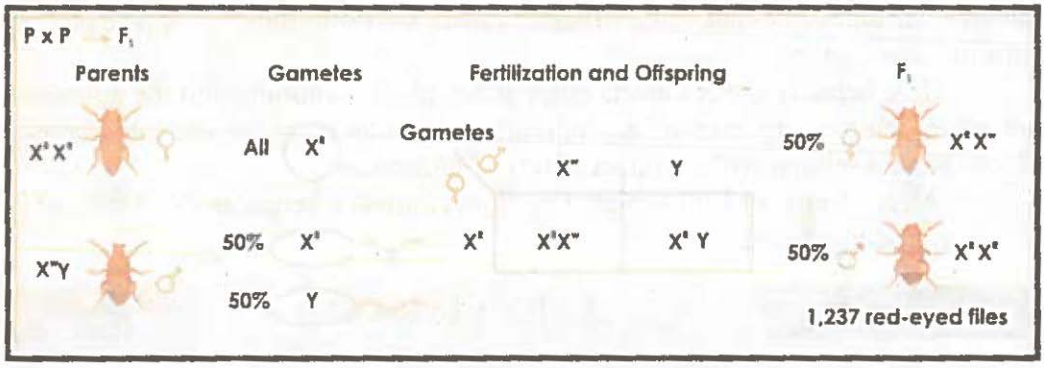


Fig: 22.13 In Morgan's first preliminary experiment (PxP), homozygous red-eyed females were mated with the white-eyed males. The female could produce gametes containing only X^R. The sperm of the male could contain either a Y chromosome or an X^w. A Punnett square is used to describe the offspring produce by the fertilization of parental gametes. The phenotypes and genotypes and ratios of the F_1 are also shown.

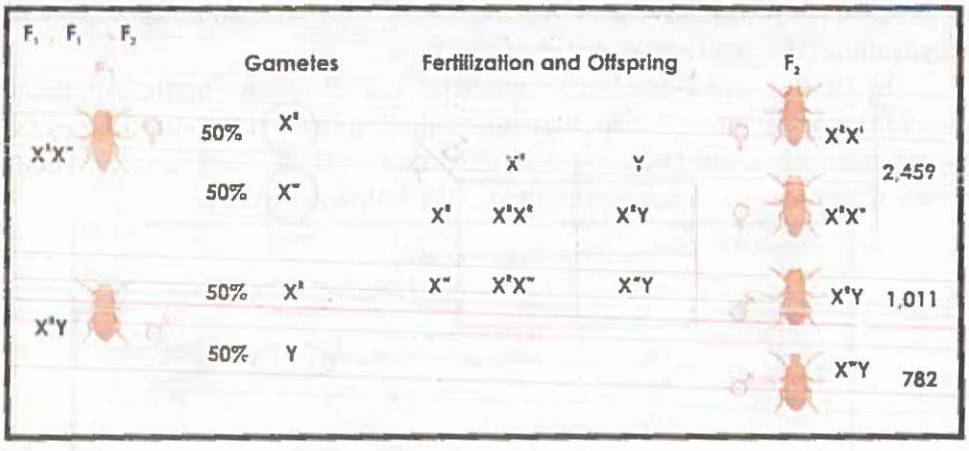


Fig 22.14 In Morgan's second preliminary experiment, F_1 females were mated with F_1 males. Morgan hypothesis explained clearly why all the white eyed flies in F_2 generation were only males.

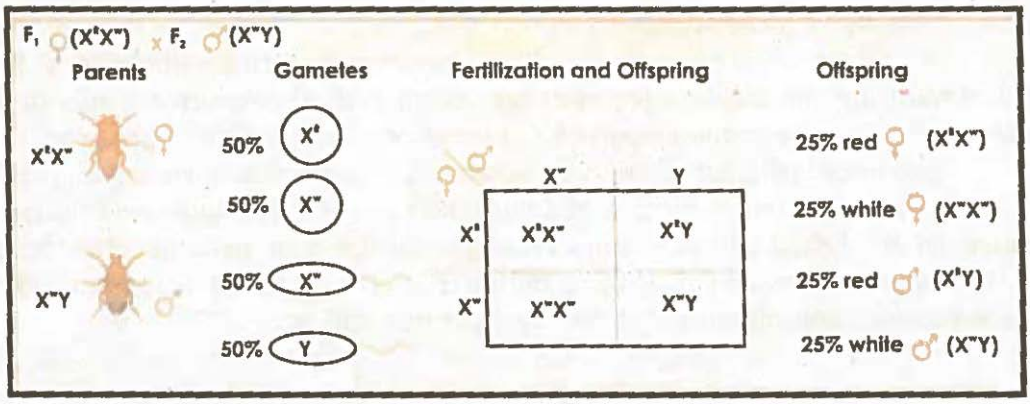


Fig 22.15 Morgan wanted to test his hypothesis .he crossed F_1 females heterozygous for white-eyes with white-eyed male. The four combinations of offspring resulted (25 percent red-eyed females, 25 percent white-eyed females, 25 percent red-eyed males and 25 percent white-eyed males).White eyed female provided an opportunity for a further confirmatory test .For this a cross was made between white eyed female (X^wX^w) with a red eyed male (X^RY). All the females' offspring's had red eyes and all male offspring had white eyes (Fig: 22.16).

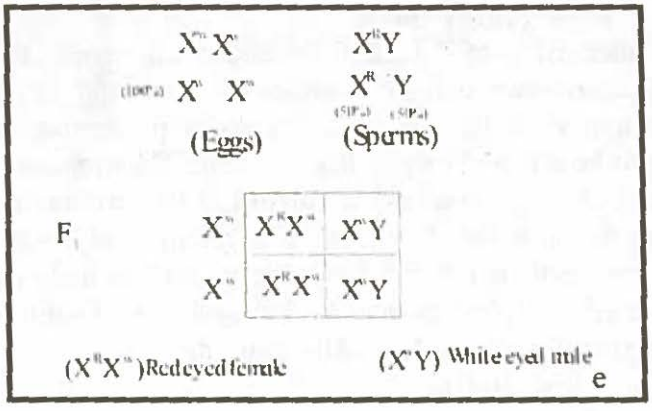


Fig: 22.16

Morgan experiments were designed to test prediction derived from his hypothesis that factor for eye colour is associated with X chromosomes. A trait whose gene is present on X-chromosome is called X-linked trait and a gene which is present on X- chromosomes with no counter part on Y –chromosome is called X-linked gene. Same is the case with Y –linked genes. Y-chromosome do carry some gene having no counter part on X chromosome e.g SRY genes on Y chromosome of man determines maleness. Y-linked genes directly pass through Y chromosome from father to son.

22.8.3 Types of sex linked traits:

A trait whose gene is present on X chromosome is called X – linkage trait. X – linked traits are commonly referred as sex-linked trait. A gene present only on X chromosome, having no counterpart on Y chromosome, is called X – linked gene.

Sex-linked inheritance follows a very specific pattern. As a son inherits his X chromosome only from his mother, and a daughter gets as X chromosome from each parent, an X – linked trait passes in a crisscross fashion from maternal grandfather (P1) through his daughter (F1) to the grandson (F2). It never passes direct from father to son because a son inherits only Y chromosome from father.

22.8.4 Sex – Linkage in Humans

Humans have many X – linked traits of which some like haemophilia and colour blindness are recessive while others like hypophosphatemic or vitamin D resistant rickets are dominant. X – linked dominant is a trait which is determined by an X linked dominant gene, while X – linked recessive is a trait that is determined by as X – linked recessive gene. Their patterns of inheritance are very different from each other.

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

X-Linked Recessive Inheritance

Characteristics of X-Linked Recessive Inheritance: Females possessing one X-linked recessive mutation are considered carriers and will generally not manifest clinical symptoms of the disorder. All males possessing an X-linked recessive mutation will be affected (males have a single X-chromosome and therefore have only one copy of X-linked genes). All offspring of a carrier female have a 50% chance of inheriting the mutation. All female children of an affected father will be carriers (daughters posses their fathers' X-chromosome). No male children of an affected father will be affected (sons do not inherit their fathers' X-chromosome).

Some examples of X-linked Recessive Disorders:

- Hemophilia A and B
- Colour Blindness
- Diabetes Insipidus

X-Linked Dominant Inheritance:

Characteristics of X-Linked Dominant Inheritance: A male or female child of an affected mother has a 50% chance of inheriting the mutation and thus being affected with the disorder. All female children of an affected father will be affected (daughters possess their fathers' X-chromosome). No male children of an affected father will be affected (sons do not inherit their fathers' X-chromosome).

Some examples of X-Linked Dominant disorders:

- Alport's syndrome
- Coffin - Lowry syndrome (CLS)
- Idiopathic hypoparathyroidism
- Vitamin D resistant rickets

Y-linked inheritance:

In mammals, Y-linkage refers to when a phenotypic trait is determined by an allele (or gene) on the Y chromosome. It is also known as **holandric inheritance.**

The Y-chromosome is small and does not contain many genes, therefore few traits are Y-linked, and Y-linked diseases are rare. Because the only humans which have a Y chromosome are males, the genes are simply passed from father to son, with no interchromosomal genetic recombination.

Chromosome Y deletions are a frequent genetic cause of male infertility. Another example in humans of a Y-linked trait was thought to be hairy ears (it may also be sex-limited). However, this has been discredited.

22.8.5 Genetics of Haemophilia:

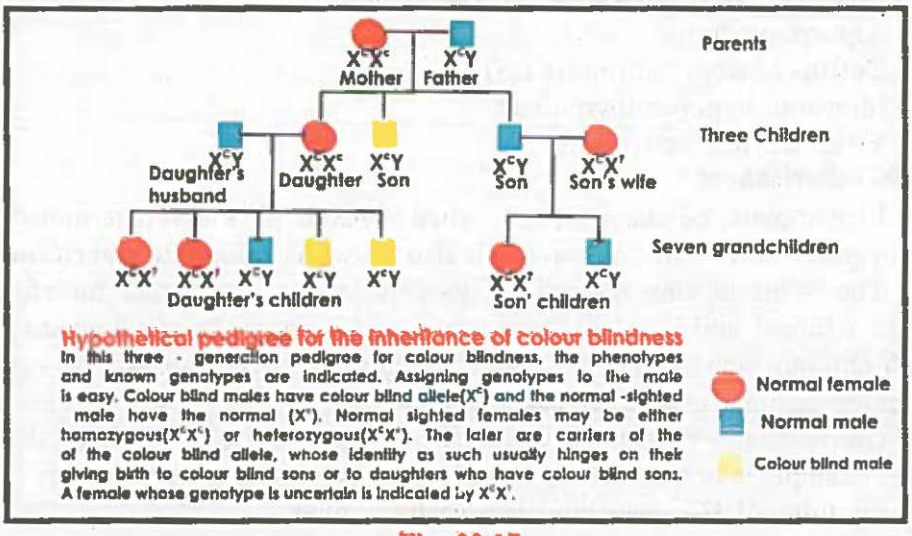
A well-studied sex-linked trait in humans is hemophilia, a disorder that renders the individual less able to form blood clots. This is a serious medical problem because in hemophiliacs, or bleeders, even a minor injury can result in a major loss of blood. Because the hemophilia allele is recessive and carried on the X chromosome, hemophilia is predominantly a male disorder. In fact, hemophilia is extremely rare in females because there are so few hemophiliac males that survive to marry and reproduce. Largely the phenotypically normal female carrier maintains the faulty allele in human population

.22.8.6 Genetics of colour-blindness:

Red green colour blindness is a recessive sex linked trait that renders individuals unable to distinguish shades of red or green and both appear gray.

To understand why colour blindness occurs much more frequently in males, let us examine the types of parental combinations that can produce colour blindness in sons and daughters.

A son inherits the colour- blind allele from his mother, who may be either colour- blind herself or, more likely, a normal- sighted carrier. There is a 50% chance that a son will inherit the colour blind trait from a carrier mother. Whether or not the father is colour-blind (X^cY or X^cY) has no bearing because a son receives only the Y chromosome from his father.



Hypothetical pedigree for the inheritance of colour blindness

In this three - generation pedigree for colour blindness, the phenotypes and known genotypes are indicated. Assigning genotypes to the male is easy. Colour blind males have colour blind allele(X^c) and the normal -sighted male have the normal (X^+). Normal sighted females may be either homozygous(X^+X^+) or heterozygous(X^cX^+). The later are carriers of the of the colour blind allele, whose identity as such usually hinges on their giving birth to colour blind sons or to daughters who have colour blind sons. A female whose genotype is uncertain is indicated by X^+X'.

Fig: 22.17

For a daughter to be colour-blind, however, not only her mother must have at least one allele for colour blindness, but her father must be colour-blind. As this combination of parents occurs rather infrequently, there are few colour-blind females in the human population.

22.8.7 Sex Related Traits:

Sex related traits are those which are associated with maleness or femaleness. These traits are not necessarily being sex linked. These traits may be controlled by sex linked or autosomal genes.

Sex limited traits:

A sex limited trait is a type of sex related trait which is confined to only one sex due to anatomical differences. Such traits affect a structure or function of the body present in only males or in only female.

For example: Genes for milk yield in dairy cattle affect only cows. Similarly beard growth in human is limited to men. A woman does not grow a beard herself but she can pass the genes specifying heavy beard growth to her son.

Sex influenced trait:

Sex influenced traits are also the type of sex related traits. These occur in both males and females but they are more common in one sex. Such traits are controlled by an allele that is expressed as dominant in one but recessive in the other. This difference in expression is due to hormonal difference between the sexes.

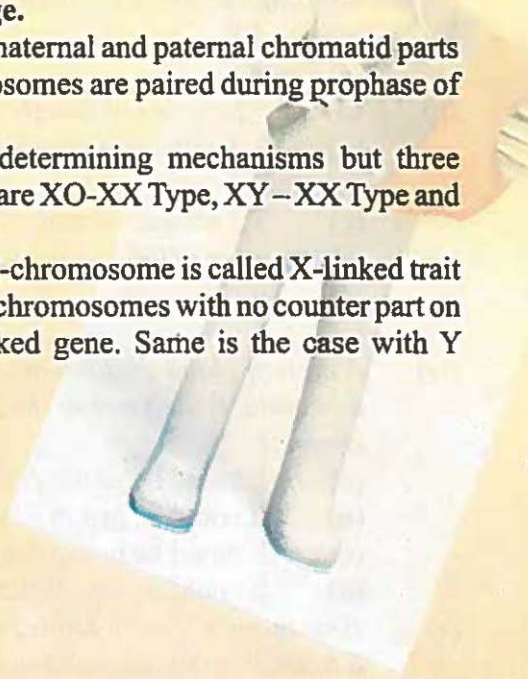
For example: Baldness is a sex influenced trait. Many more men than women are bald. It is inherited as an autosomal dominant trait in males but as an autosomal recessive trait in females. A heterozygous male is bald but a heterozygous female is not. A woman can be bald only when she is homozygous recessive.

KEY POINTS

- The tendency of individuals to resemble their parents is known as **heredity**.
- The science which deals with mechanism of the heredity and variation is called **genetics.**
- Mendel was the first who successfully explained the mechanism of inheritance during his research work on pea plant.
- According to the law of dominance, different characters are controlled by units called factors; factors occur in pairs, of a pair, one factor dominates the other.
- A dihybrid cross describes a mating experiment between two organisms that are particularly different for two traits. The offspring of such cross is called dihybrid which is heterozygous at two different genetic loci.
- Overall, dominance relations are: complete dominance, incomplete dominance, co-dominance and over dominance.
- ABO blood groups are found in all humans and in many other primates such as apes chimpanzees, baboons, and gorillas. There are four principal types: A, B, AB, and O, there are two antigens and two antibodies that are mostly responsible for the ABO types.
- The Rh blood group system currently consists of 50 defined blood-group antigens, among which the 5 antigens D, C, c, E, and e are the most important ones.
- **Epistasis is** defined as the phenomenon in which the effect caused by the genes at one locus interferes with or hides the effect caused by another gene at another locus.
- 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**. These quantitative traits are **polygenic traits.** All the genes that control a quantitative trait are called **polygenes** which have a small positive or negative effect on the character.
- Human skin colour is a good example of polygenic (multiple gene) inheritance.

KEY POINTS

- The phenomenon of staying together of more than one gene on the same chromosome is called **gene linkage.**
- In crossing over, an exchange of maternal and paternal chromatid parts occurs while homologous chromosomes are paired during prophase of meiosis I.
- There are wide variety of sex determining mechanisms but three patterns are more common which are XO-XX Type, XY – XX Type and XX – XY type or WZ – ZZ Type.
- A trait whose gene is present on X-chromosome is called X-linked trait and a gene which is present on X- chromosomes with no counter part on Y –chromosome is called X-linked gene. Same is the case with Y –linked genes.



EXERCISE ?

1- Multiple Choice Questions

(i) ***Blood group B phenotype contains anti-A antibodies in the serum and agglutinates any RBC with antigen:***

(a) AB (b) O
(c) A (d) B

(ii) ***Chances for a son or daughter in human birth are:***

(a) 3:1 between son and daughter (b) 1:3 between son and daughter
(c) 1:1 between son and daughter (d) None of them

(iii) ***The number of linkage group in man is:***

(a) 02 (b) 23
(c) 46 (d) 92

(iv) ***A man of blood group A marries a woman of blood group B and they have one child. Which one of the following statements about the child's blood is correct?***

(a) It could be group A only
(b) It could be group AB only
(c) It could be group A or Group B only
(d) It could be any of the groups A, B, AB, O.

(v) ***How many different kinds of gametes will be formed by an individual, who is heterozygous for four gene pairs:***

(a) 8 (b) 16
(c) 20 (d) 30

(vi) ***A woman with normal colour vision, whose father was red-green colour blind, married a red-green colour-blind man. What is the probability of her first-born child being red-green colour blind?***

(a) 1.0 (b) 0.75
(c) 0.50 (d) 0.025

(vii) ***Two parents, each of blood groups A, have a daughter of blood group O. What is the probability that their next child who has blood group O?***

(a) 0.125 (b) 0.25
(c) 0.50 (d) 0.75

(viii) ***What are the phenotypes of the parent of a colour-blind son and non-carrier daughter with normal colour vision?***

EXERCISE ?

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

(ix) ***When expression of a biological character is observed in variable intensity it is due to the affect of:***

(a) multiple alleles (b) codominance
(c) epistasis (d) polygenic inheritance

(x) ***Inheritance of skin colour in man is controlled by eight pairs of genes, which are:***

(a) linked (b) codominent
(c) multiple alleles (d) assorting independently

2- Short Questions

(i) Why pea was the most suitable plant for Mendel's experiments.
(ii) Differentiate between dominance and epistasis.
(iii) State the dominance relations among the alleles of ABO blood group system
(iv) What were the Mendel's observations about the phenotypes and genotypes in F_2 generation of monohybrid cross.
(v) Differentiate between sex limited and sex influenced traits

3- Long Questions

(i) Define and explain the law of segregation.
(ii) How Mendel performed dihybrid cross? What he did proposed on the basis of his observations in this cross.
(iii) Describe the genetic basis of ABO blood group system.
(iv) What is polygenic inheritance? Explain its mode of inheritance by taking an example in human.
(v) Describe Morgan's historic work on sex linkage in Drosophila.

4- Genetic Problems

(i) A woman with Type O blood and a man with Type AB are expecting a child. What are the possible blood types of the kid?

EXERCISE ?

(ii) What are the possible blood types of a child whose parents are both heterozygous for "B" blood type?

5- **Analyzing and Interpreting**

- Collect data from class or the school to see how many individuals have AB blood group and construct a pie chart and histogram for collected data.

6- **Initiating and Planning**

- Hypothesize that in dihybrid inheritance pattern of colour and texture of pea seed, the two traits are not inter dependent.

7- **Performing and Recording**

- Build a thematic chart for blood groups for his/her class fellows and identify the antigens present in blood.
- Test his/her blood group using antisera and explain which antigens and antibodies he/she has?

8- **Science, Technology, and Society Connections**

- Evaluate incomplete and co-dominance as variation of Mendel's research.
- Drive an idea to get alternative of blood transfusion. (reference could be made to synthesize plasma and serum)
- Justify why thc recessive blood group allele ""i" is more frequcnt in the population.
- Justify blood donation as a service to suffering humanity.
- Name and explain the techniques employed for embryonic screening, e.g. Amniocentasis.
- Suggest ways to solve lives through the knowledge gained in this chapter.
- Describe how the field of genetics has progressed to more applied science.
- Justify the effectiveness of some of the treatments of haemophilia.

6- **Online Learning**

- *www.phschool.com/science/biology*
- *www.ncbi.nlm.nih.gov*
- *www.nature.com*
- biology.clc.uc.edu/courses/bio105/geneprob.htm
- www.sciencedaily.com