Chapter 18

INHERITANCE

STUDENTS' LEARNING OUTCOMES

After studying this chapter, the students will be able to:

- Define gene.
- Explain the law of segregation and independent assortment, using a suitable example related to the pea plants.
- Relate the Law of independent assortment to random orientation of chromosomes during meiosis.
- Express limitations of independent assortment and its usefulness.
- Show that independent assortment leads to variation in the gametes.
- Evaluate that inheritance of genes and their mixing during fertilization is based on mathematical probabilities.
- Describe the exceptions to the Mendel's laws of inheritance.
- Explain incomplete dominance and exemplify it through the inheritance of flower color in 4 O' clock plant.
- Differentiate between incomplete dominance and codominance.
- Define alleles and multiple alleles.
- State the alleles responsible for the trait of ABO blood groups.
- Explain the case where two alleles have equal dominance through the genetics of human blood group AB.
- Name the various human blood group systems.
- Investigate the reasons for O-ve individual as the Universal donor and AB +ve as the Universal recipient.
- Describe the occurrence of some other blood group systems.
- Associate the positive and negative blood groups with the presence and absence of Rh factor.
- Justify why Rh incompatibility could be a danger to the developing fetus and mother.
- Explain Erythroblastosis fetalis in the light of antigen-antibody reaction.
- Suggest measures to counter the problem of Erythroblastosis fetalis before it occurs.
- Define and relate the terms; polygenic and epistasis.
- Describe polygenic inheritance using suitable examples from plants (grain color in wheat) and animals (skin color in man).
- List at least five polygenic traits discovered in humans.
- Give one example of epistasis from mammals (coat color inheritance in Labrador retrievers) and one from plants (pigment phenotype in foxgloves) and justify modified Mendelian ratios.
- Describe the terms gene linkage and crossing over.
- Explain that gene linkage counters independent assortment and crossing-over modifies the progeny.

- Suggest that linkage can be observed / evaluated only if the number of progeny is quite large.
- Explain the XX-XY mechanism of sex determination in mammals.
- Identify male and female individuals from the karyotype of man.
- Solve the genetics problems related to XX-XY, sex determination.
- Describe the concept of sex-linkage.
- Explain the inheritance of sex-linked traits (eye color) in Drosophila.
- Describe the sex-linked inheritance of male characters due to Y-chromosome and the effect of Holandric genes.
- Describe the X-linked disorders with reference to the patterns of inheritance.
- Name some of the sex-linked disorders of man (Red green color blindness, Hemophilia) .
- Explain the techniques employed for embryonic screening e.g., Amniocentesis and Chorionic Villus Sampling.

You have studied the basics of Mendelian genetics in Grade X. This chapter carries the concept forward to post-mendelian research. It also gives an insight into the inherited diseases and their symptoms and treatment.

Recalling
The Austrian monk, Gregor Johann Mendel, was the first to explain the mechanism of inheritance. He developed true-breeding (pure-breeding) varieties of pea plants. A plant which is true-breeding for a particular trait produces all the offspring with the same trait upon self-fertilization.

18.1- Law of Segregation

Mendel performed monohybrid crossed in pea plant. In the first monohybrid crosses, he observed the inheritance of "seed shape". When he crossed a true-breeding round-seeded plant with wrinkled-seeded plant, all plants of F1 generation produced round seeds. He called the trait that expressed in the F1 plants (round seeds) as **dominant** and the alternative trait that was not expressed in the F1 plants (wrinkled seeds) as **recessive** trait.

When F1 plants were self-pollinated, the 1/4 of F2 seed were wrinkled. In other words, the dominant-recessive ratio among the F2 seeds was 3 dominant : 1 recessive. His results suggested that the 3:1 ratio in F2 generation was really a disguised 1:2:1 ratio.

In F2 generation,

- 1⁄4 were pure-breeding dominant
- 2⁄4 were not pure-breeding dominant
- 1⁄4 were pure-breeding recessive

From his experiments, Mendel concluded that for each pair of alternative traits, one trait was not expressed in the F1 hybrids, although it reappeared in 1/4 of F2 seeds. The trait that "disappeared" must therefore be disguised in the F1 seeds.

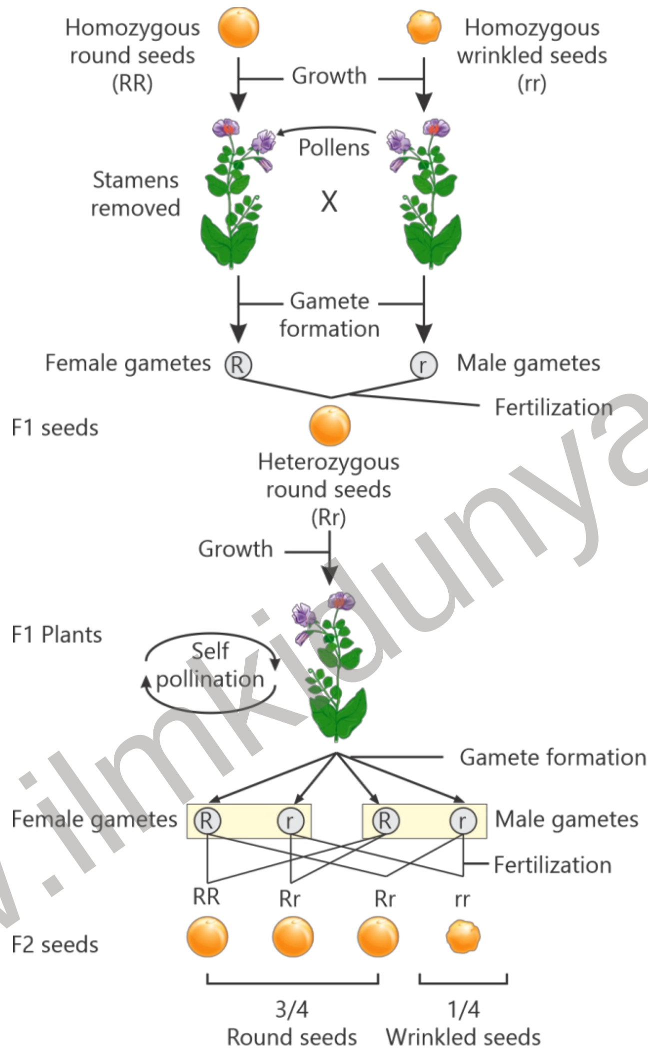


Figure 18.1: Mendel's monohybrid cross

Interpretation of the Results

Mendel explained that the characters (traits) of organisms are controlled by special "factors" (which are now called **genes**). Each individual is diploid and has two genes for a character. Gene are present on chromosomes. When the individual forms haploid gametes (eggs or sperms), each gamete receives one gene for each character.

All copies of a gene may not be identical. The alternative forms of a gene, leading to alternative traits, are called **alleles**. When two haploid gametes containing the same alleles fuse, the offspring receives the gene-pair with similar alleles. Such offspring is called **homozygous** for that gene pair or trait. When the two haploid gametes with different alleles fuse, the offspring is **heterozygous**.

We now know that a gene is **"a sequence of nucleotides as part of DNA, which codes for the formation of a polypeptide"**. The particular location of a gene on a chromosome is called the gene's locus.

When the individual forms gametes, each gamete receives just one gene copy (allele), which is selected randomly. This is known as the law of segregation. When gametes join, they form a new individual, whose genotype consists of the alleles contained in the gametes.

For Information

In heterozygous individuals, only one allele (dominant) is expressed, while the other allele (recessive) is present but unexpressed.

The combination of alleles in an individual is its **genotype**. The **phenotype** is the observable character or trait. In other words, the genotype is the blueprint, and the phenotype is the visible outcome.

Testcross

A testcross is used to determine whether a dominant phenotype is homozygous or heterozygous. It involves crossing the dominant phenotype with a recessive phenotype. For example;

A round-seeded plant may be RR (homozygous) or Rr (heterozygous), while wrinkled-seeded plants are always rr. To determine its genotype, the round-seeded plant is crossed with a wrinkled-seeded plant (rr).

- If it is **RR**, all offspring will be **round (Rr)**.
- If it is **Rr**, the offspring will be **round and wrinkled in a 1:1 ratio**.

18.2- Law of Independent Assortment

After showing that alleles of one character segregate independently, Mendel studied whether genes of different characters also segregate independently. He examined two characters together: seed shape — round (R) dominant and wrinkled (r) recessive, and seed colour — yellow (Y) dominant and green (y) recessive. He established pure-breeding lines for these traits.

1. Pure-breeding plants with both dominant traits i.e., round and yellow seeds (RRYY)
2. Pure-breeding plants with both recessive traits i.e., wrinkled and green seeds (rryy)

He crossed pure-breeding plants (**RRYY** × **rryy**). All F1 seeds showed both dominant traits (round and yellow) and were heterozygous with genotype **RrYy**. These F1 plants are called **dihybrids**, as they are heterozygous for both genes.

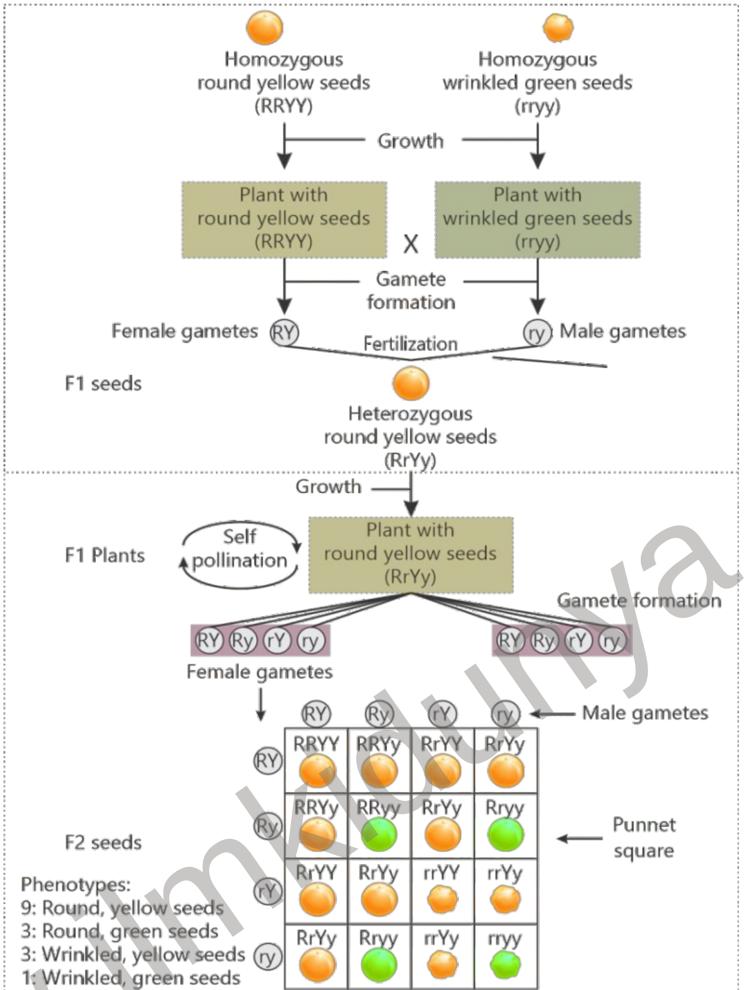


Figure 18.2: Mendel's dihybrid cross

To confirm the genotype of F1 plants, Mendel self-fertilized them to produce the F2 generation. Instead of a 3:1 ratio, the F2 showed four phenotypes i.e., round yellow, round green, wrinkled yellow, and wrinkled green - in a **9:3:3:1 ratio**.

> **For Information**
> Independent assortment of different genes does not affect the segregation of individual allele pairs. Thus, round vs wrinkled seeds show a 3:1 ratio, and yellow vs green seeds also show a 3:1 ratio.

Interpretations of the Results

In a F1 dihybrid plant (**RrYy**), seed shape has alleles **R, r** and seed colour has **Y, y**. During gamete formation, four combinations are produced: **RY, Ry, rY, ry**. When these gametes combine (4 male × 4 female gametes), **16 combinations** are formed in the F2 generation. This results in four phenotypes in F2 in a **9:3:3:1 ratio**:

- 9 round yellow
- 3 round green
- 3 wrinkled yellow
- 1 wrinkled green

The results showed that genes for seed shape assort independently of genes for seed colour. On these basis, Mendel proposed that "each pair of alleles assorts independently of other pairs of alleles during gamete formation". It is referred to as Mendel's law of independent assortment.

Independent Assortment as a Source of Variations

Independent assortment of alleles creates genetic diversity. It mixes alleles during the formation of gametes (sperm and egg cells). Durin gamete formation, alleles for different traits, which are on different chromosomes, separate independently of each other. This means one character (like eye colour) does not affect another character (like hair texture).

Tidbit
A modern re-statement of Mendel's law of independent assortment would be that *"genes that are located on different chromosomes assort independently during meiosis".*

Independent assortment happens because homologous chromosome pairs line up randomly during meiosis. This random alignment causes different combinations of maternal and paternal alleles in gametes. In humans, it can produce over 8 million (2^{23}) possible allele combinations in one gamete. This reshuffling makes every gamete genetically unique.

The chances of variation increases further during fertilization. When a sperm and egg fuse, they combine their unique alleles to form a genetically unique zygote. This continuous mixing of alleles in a population creates genetic diversity, which helps species adapt to changing environments.

18.2.1- Limitations to the Law of Independent Assortment

The law of independent assortment helps explain how genes are inherited, but it does not cover all genetic complexities. For example;

1. **Linked genes:** Genes close together on the same chromosome are often inherited together, not independently. This phenomenon is known as **gene linkage**.
2. **Chromosome crossover:** It is the exchange of genetic material between homologous chromosomes during meiosis. Due to this process, the ratio of gene combinations may not be as predicted by the law of independent assortment.
3. **Epistasis:** It is phenomenon where the effect of one gene is modified or suppressed by one or more unrelated genes Epistasis also changes the expected ratio of gene combinations.

4. **Pleiotropy:** It is the phenomenon where one gene controls multiple characters. It causes the characters to be linked and controlled by a single gene. Pleiotropy can distort the expected phenotypic ratios from independent assortment.
5. **Environmental effect:** Many phenotypes can be affected by the environment.

18.2.2- Inheritance and Mathematical Probabilities

The transfer of genes from one generation to the other is random. The calculation of the possible new allelic combinations is based on mathematical probability. Probability means how often a particular outcome occurs.

Probability of inheritance of one or two alleles (e.g., RrYy × RrYy) can be calculated using a Punnett square. However, for many genes (e.g., AaBbCcDdEe × AaBbCcDdEe), Punnett squares become too complex. So, probability rules are used instead.

Basic Rule of Probability

Probability is calculated by dividing the number of times an event occurs by the total number of possible outcomes.

For example, if wrinkled pea seeds appear 1,850 times out of 7,324 seeds, the probability is 1,850 ÷ 7,324 = 0.253, which is about 1 in 4 seeds.

> **Performing and Recoding**
> Use the rules of probability and calculate the probability of getting Sixes when you roll a dice 100 times.

The Product Rule of Probability

The product rule of probability says that the chance of two or more independent events happening together is calculated by multiplying their individual probabilities. For example, getting a six on one dice is 1/6. For two dice, the chance of getting two sixes is (1/6) × (1/6) = 1/36.

The product rule can be used to predict fertilization outcomes.

Example 1 (Aa × Aa):

- Each parent has a 1/2 chance of giving gamete with allele *a*.
- So, probability of aa offspring = (1/2) × (1/2) = 1/4.

Example 2 (RrYy × RrYy):

- Probability of Round seed = 3/4, Probability of yellow seed = 3/4.
- So, probability of round yellow offspring = (3/4) × (3/4) = 9/16.

The Sum Rule of Probability

This rule applies to mutually exclusive events (only one can happen at a time). The probability of either event is found by adding event's individual probabilities. For example, on a six-sided dice, the chance of getting 1 is 1/6 and the chance of getting 6 is 1/6. So, getting either 1 or 6 will be (1/6) + (1/6) = 1/3.

Example

In a cross **Aa × Aa**, dominant phenotype (AA or Aa) can occur in three ways:

- A from male + A from female (AA)

- A from male + a from female (Aa)
- a from male + A from female (Aa)

Each individual event has a probability of **1/4**.

So, total probability of dominant phenotype = (1/4) + (1/4) + (1/4) = **3/4**.

Analyzing, Interpreting and Communication

Solve the following genetic problems.

- Phenylketonuria and albinism are two recessive disorders which assort independently. If a mother and a father who both are heterozygous for both traits, produce a child, what is the chance of their having a child with (a) phenylketonuria, (b) albinism, and (c) both traits?
- A gene has two alleles i.e., dominant C and recessive c. What proportions of the offspring from a CC x Cc cross would be homozygous dominant, homozygous recessive and heterozygous?
- A TtYy pea plant self-pollinates and one seed is picked at random for planting. (a) What is the chance that the seed will produce a tall, green seeded plant? (b) If it turns out to be tall and yellow-seeded what is the chance that its genotype is TTYY?

18.3- Dominance Relations and Multiple Alleles

In pea plants (Yy), the dominant allele (Y) is fully expressed and the recessive allele (y) is not expressed at all. So no intermediate trait (green) appears in Yy. This is called **complete dominance**. Scientists tested Mendel's ideas and found that results were not always the same. Many traits do not show complete dominance. Moreover, some traits are controlled by more than two alleles.

18.3.1- Incomplete Dominance

In **incomplete dominance**, neither allele is fully dominant over the other. In such situations, the heterozygous individual shows a phenotype that is intermediate between phenotypes of both parents. For example, in Japanese four-o'clock plants, when a red flowered (RR) is crossed with white flowered (WW), it produces all heterozygous pink flowered (RW) offspring. This shows neither the alleles for red (R) nor of white (W) is completely dominant. When two pink flowered

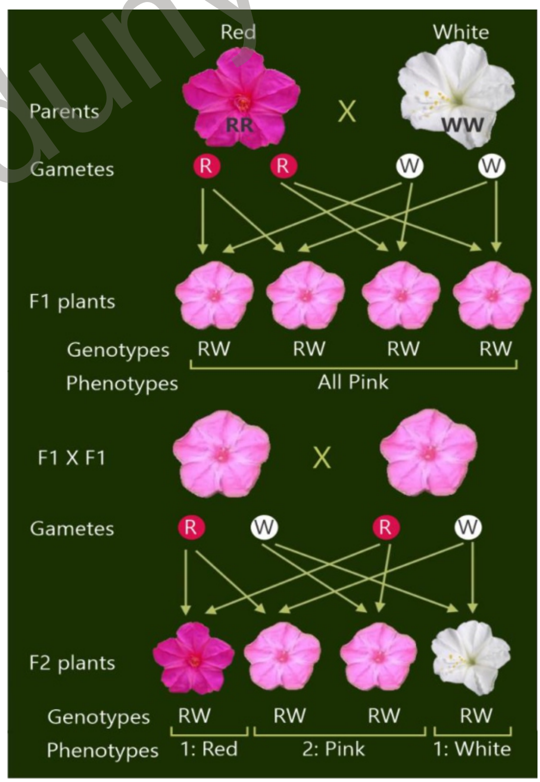


Figure 18.3:Incomplete dominance in Japanese 4-o'clock plants

plants (RW × RW) are crossed, the result is red (RR), pink (RW), and white (WW) in a **1:2:1 ratio**.

Solve the following genetic problem:
A pink flower four O'clock plant is crossed with a red flower plant. Find:
(a) Probability of Red flower plant
(b) Probability of pink flower plant
(c) Ratio of pink flower to red flower plant

18.3.2- Co-dominance

Co-dominance is type of inheritance in which both alleles in a gene pair are fully expressed in a heterozygous individual. In co-dominance, both alleles work without blending or one masking the other. So, the products of both alleles appear together in the phenotype of heterozygous.

For Information
The "L" stands for Landsteiner and Levine, the scientists who discovered this system in 1927.

For Information
It is also called the MNS system because the genes of MN group and the S/s group are physically "linked" on the same chromosome.

The human **MN blood group** shows codominance, where both alleles are equally expressed. There are two alleles for this system.

- Allele L^M Produces M antigen.
- Allele L^N Produces N antigen.

Both L^M and L^N are codominant. So, if both are present, both antigens appear on red blood cells. In this way, there are three possibilities i.e.,

1. M blood group (L^ML^M) – individuals with only M antigens.
2. N blood group (L^NL^N) – individuals with only N antigens.
3. MN blood group (L^ML^N) – individuals with both M and N antigens.

MN Blood Group System

Genotype	Antigen	Blood Group Phenotype
L^ML^M	M	M blood group
L^NL^N	N	N blood group
L^ML^N	M & N	MN blood group

Table: Comparison between Incomplete Dominance and Codominance

Incomplete dominance	Co-dominance
1. In heterozygous state, both genes blend their phenotypic effects.	1. In heterozygous, both genes fully express their effects.
2. The heterozygous show an intermediate phenotype between the two parental phenotypes.	2. The heterozygous show both parental phenotypes at a time.

3. Example: Flower colour in Japanese 4 O' clock plant	3. Example: Human MN blood group and human AB blood group

18.3.3- Multiple Alleles

A **multiple allelic system** occurs when a gene has more than two alternative forms (alleles) in a population. This creates more variation in traits.

An example is the human **ABO blood group system**. It is controlled by one gene (I gene) with three alleles: **I^A, I^B, and i**. Each person has only two of these alleles. Different combinations produce four blood groups:

Phenotypes (Blood groups)	Genotypes
1. **A**	I^AI^A or I^Ai
2. **B**	I^BI^B or I^Bi
3. **AB**	I^AI^B
4. **O**	ii

For Information
In human ABO blood group system, the alleles I^A and I^B also show co-dominance.

18.4- Blood Group Systems

A blood group system is a way of classifying blood based on the presence of specific antigens on red blood cells (RBCs). The International Society of Blood Transfusion recognizes **43 blood group systems** in humans. Each system has different blood groups.

For Information
In blood group systems, the antigens present on the surface of RBCs may be proteins, carbohydrates, glycoproteins, or glycolipids.

The most important is the **ABO system** (blood groups A, B, AB, O), and the second is the **Rh system** (blood groups Rh-positive and Rh-negative). For blood transfusion, the matching of ABO and Rh-system is necessary between donor and recipient's blood. The following are the other major blood group system, but these usually do not cause matching problems in the blood transfusions.

Blood Group System	Well-known antigens
MNS	M, N, S, s, U
Lutheran	Lu^a and Lu^b
Kell	K, k, Kp^a, Kp^b, Js^a, Js^b
Lewis system	Le^a and le^b
Duffy	Fy^a, Fy^b, Fy^3

18.4.1- ABO Blood Group System

The ABO system was discovered by Landsteiner in 1901. He found two antigens on red blood cells (RBCs): **antigen-A and antigen-B**. He observed some individuals have antigen-A on their RBCs while others have antigen-B. He also observed that many individuals do not have any of these antigens. Later, Landsteiner's students discovered that some individuals have both A and B antigens on their RBCs. So, they named the different blood groups as:

- People with antigen-A — blood group A
- People with antigen-B — blood group B
- People with both (antigen- A & antigen-B — blood group AB
- People with neither of antigen-A & antigen-B — blood group O

Each person's blood plasma contains antibodies against the antigen s/he does not have:

- Blood group A → anti-B antibodies
- Blood group B → anti-A antibodies

This is called **Landsteiner's rule of antigen–antibody relationship**.

For Information
Some people secrete antigens A and B in body fluids e.g., saliva, tears, urine. They possess a dominant secretor gene "Se" on their chromosome 19.

Inheritance of the ABO Blood Groups

The ABO system is controlled by the gene I ("I" stands for iso-haem-agglutinogen). It is located on chromosome 9 and has multiple alleles i.e., I^A, I^B and i. The alleles I^A and I^B are co-dominant over each other. While allele i is recessive to both of them. The I^A allele produces antigen-A, the I^B allele produces antigen-B. Allele i does not produce any antigen. Therefore;

- Genotypes I^AI^A and I^Ai produce antigen-A. This phenotype is called **blood group A**. Individual's plasma contains anti-B antibodies.
- Genotypes I^BI^B and I^Bi produce antigen-B. This phenotype is called **blood group B**. Individual's plasma contains anti-A antibodies.
- Genotype I^AI^B produces both antigen-A and antigen-B. This phenotype is called **blood group AB**. Individual's plasma does not contain anti-A or anti-B antibodies.
- Genotype ii produces neither antigen-A nor antigen-B. This phenotype is called **blood group O**. Individual's plasma contains both anti-A and anti-B antibodies.

For Information
In ABO system, the production of anti-A and anti-B antibodies does not require any exposure to antigens. Rather, these antibodies start forming from embryonic life and continue throughout life.

Solve the following genetic problem.

- A person has blood group A while his wife has B. They have four children each with different blood group. Explain that how is it possible?
- A woman with blood group B has a child with blood group O. What are the genotypes of the mother and child?

For Information
In pea plants, each character that Mendel studied was controlled by a gene with two alleles. One allele was completely dominant over the other. But incomplete dominance and co-dominance do not obey this rule of dominance. Similarly, in multiple allelic system, a gene has more than two alleles,

Which genotypes could the father not have?

any two of which work for producing a phenotype.

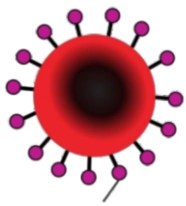
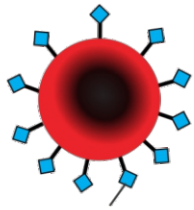
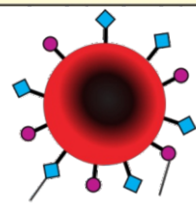
Genotypes	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Antigens (on RBCs)	 Antigen-A	 Antigen-B	 Antigen-B Antigen-A	 No Antigens
Antibodies (in Plasma)	 Anti-B	 Anti-A	No Anti-A antibody No Anti-B antibody	 Anti-A Anti-B
Phenotypes (Blood Groups)	A	B	AB	O

Figure 18.4: ABO system genotypes and phenotypes

Principle of Transfusion in ABO System

During blood transfusions between two incompatible (unmatching) blood groups, the recipient's antibodies attack the antigens on donor's RBCs. It causing **clumping (agglutination)** of RBCs. It happens;

- If blood group A individual receives blood group B and vice versa, and
- If donor's blood group is AB and the recipient is A, B, or O.

Exceptions;

- O blood group (donor): has no antigen-A and antigen-B. So, it can be given to A, B, AB, or O. So, O blood group are called **universal donors of ABO system**.
- AB blood group (recipient): has no anti-A or anti-B antibodies. So, it can receive all blood types. So, AB group are called **universal recipients of ABO system.**

Justify why a recessive blood group allele 'i' is more frequent in the population.

- In a cross between blood group O (ii) and O (ii), all offspring will have blood group O (ii).
- In a cross between heterozygous I^Ai and I^Ai, 25% offspring will have blood group O (ii)
- In a cross between heterozygous I^Bi and I^Bi, 25% offspring will have blood group O (ii).
- In a cross between heterozygous I^Ai and I^Bi, 25% offspring will have blood group O (ii).

That's why blood group allele 'i' is more frequent in the population.

18.4.2- Rh Blood Group System

It is the second most important blood group system in humans. The name of this system (Rh) is derived from **Rhesus monkey**, in which its antigen was first discovered by Landsteiner in 1930s. This system is based on the presence or absence of **Rh-antigen** (also called Rh factor or antigen-D) on RBCs. The individuals having this antigen are called **Rh-positive** and those in which it is absent are called **Rh-negative**.

Usually, the blood group of an individual mentions both ABO and Rh systems together. For instance, the individual who has antigen-A and Rh-antigen will have blood group A-positive. The individual who has antigen-A but lack Rh-antigen will have blood group A-negative. Similar is the case with individuals of blood groups B, AB and O.

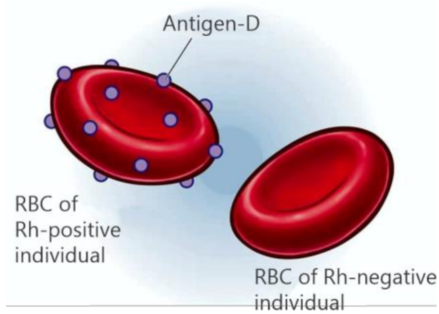


Figure 18.5: The Rh-blood group system

Inheritance of Rh Blood Groups

Rh blood group system is controlled a gene "D". It has two alleles (D and d). The dominant allele D forms the Rh-antigen, while the recessive allele d inhibits the formation of Rh-antigen. Therefore, individuals with genotypes DD or Dd have Rh-antigen and are Rh-positive. While the individuals with genotype dd do not have Rh-antigen and are Rh-negative.

Principle of Transfusion in Rh System

In ABO blood groups, the anti-A or anti-B antibodies are already present in plasma. But in Rh system, Rh-negative people do not naturally have anti-Rh antibodies. But, if Rh-negative person receives Rh-positive blood, his body start producing anti-Rh antibodies, which destroy the donated RBCs. These antibodies stay in the body for life.

The blood group O-negative is the **actual universal donor** because neither has any antigen of ABO system nor has antigen of Rh system. The blood group AB-positive is the **actual universal recipient** because it has neither anti-A and anti-B nor anti-Rh antibodies. Therefore, it receives any donor's blood.

18.4.3- Erythroblastosis Foetalis

Erythroblastosis foetalis is a **haemolytic disease** of the foetus/new-born caused by **Rh incompatibility** between Rh-negative pregnant mother and Rh-positive foetus.

Reason and Complications

It occurs when Rh-negative mother (dd) carries Rh-positive baby (DD or Dd). Foetal blood may enter the mother's blood. On exposure to Rh-antigens of foetus, mother will produce anti-Rh antibodies. These antibodies can cross the placenta and enter foetus blood. Here, mother's and anti-Rh antibodies will destroy foetal RBCs, leading to **haemolytic anaemia** and other serious complications in foetus.

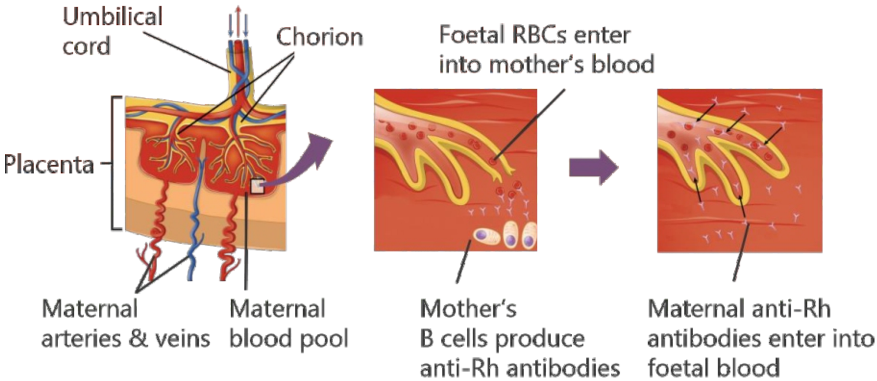


Figure 18.6: Erythroblastosis foetalis

In severe cases, destruction of foetal RBCs can cause **miscarriage or stillbirth**. If the foetus survives, the liver and spleen become enlarged because they try to make more RBCs. The breakdown of RBCs produces bilirubin, which builds up and causes jaundice (yellow skin and eyes) in foetus. High bilirubin can also damage brain cells. The surviving baby's blood is replaced by Rh-negative blood free of anti-Rh antibodies.

Tidbit
Sometimes ABO incompatibility protects the baby against Rh incompatibility. If O-negative mother conceives A-positive or B-positive baby, any foetal RBCs entering the mother's blood are quickly destroyed by her already present anti-A or anti-B antibodies before she can form anti-Rh antibodies.

Tidbit
When there are chances of Rh-incompatibility, the mother is given an injection of Rh-antiserum (serum containing anti-Rh antibodies) during early pregnancy and immediately after birth. This causes the destruction of any of the baby's RBCs that may have crossed into mother's blood before sensitizing the mother's immune system.

Solve the following genetic problem.
An Rh-negative woman is married to an Rh-positive man, whose father was also Rh-negative. What are the possible genotypes of each person in the family, what are the chances that their child will be affected with erythroblastosis foetalis?

18.5- Polygenic Inheritance and Epistasis

Polygenic inheritance and epistasis are further examples of deviation from Mendel's inheritance.

18.5.1- Polygenic Inheritance

Polygenic characters are controlled by many gene pairs, not just one. In polygenic inheritance, all alleles of many genes contribute to the same trait in an **additive way** (some increase, some decrease the effect). The combined effect produces a single trait.

Polygenic traits show a range of differences i.e., **continuous variation** in a population. Examples of polygenic traits include height, skin colour, weight, intelligence in humans, and grain colour in plants.

Inheritance of Human Skin Colour

Skin colour depends on the amount of **melanin** pigment. More melanin means darker skin; less melanin means lighter skin. This trait is controlled by polygenes (about 60 genes), but for understanding, we take 3 genes: **A, B, C**. Each of these genes has a dominant and a recessive allele.

- Alleles **A, B, and C** produce more melanin
- Alleles **a, b, and c** produce less melanin

If the parents have genotypes AABBCC (very dark) and aabbcc (very light), all of their children will be AaBbCc (intermediate colour). If both parents are triple heterozygotes i.e., AaBbCc, their children will have wide range of skin colours from very dark to very light. The ratio of different skin colours will be **1 : 6 : 15 : 20 : 15 : 6 : 1** (from very dark to very light).

- **AABBCC** → darkest (maximum melanin)
- **aabbcc** → lightest (minimum melanin)

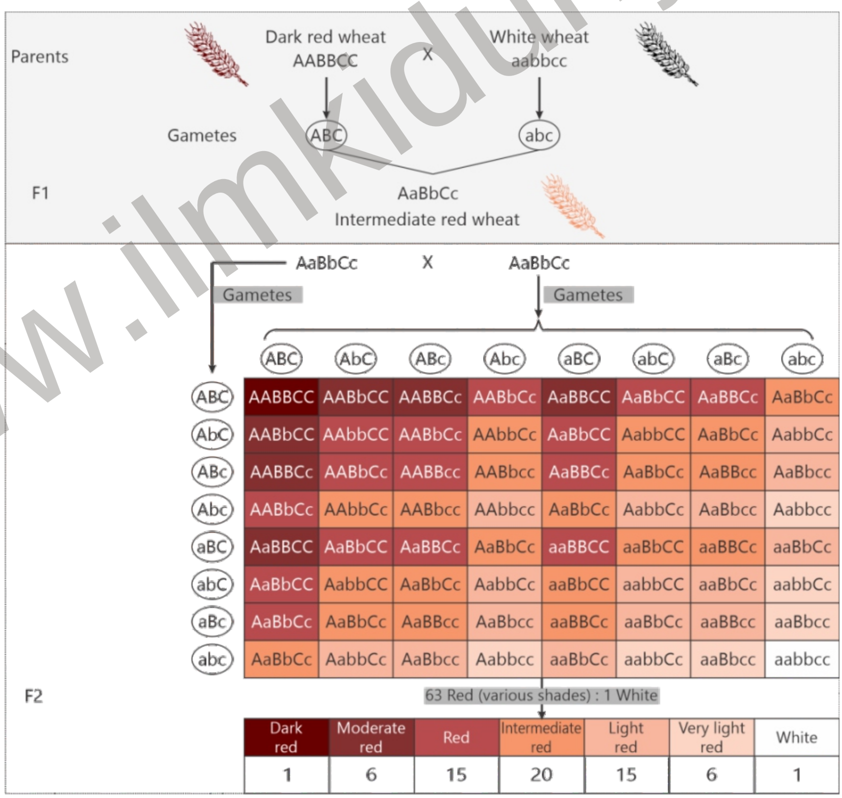


Figure 18.7: Polygenic inheritance of what grain colour

Wheat Grain Colour

Wheat grain colour shows continuous variation, ranging from white to dark red. This trait is controlled by three polygenes: A, B, and C. Each gene has a dominant allele (A, B, C) that produces red pigment and a recessive allele (a, b, c) that does not. The genotype AABBCC produces the maximum red pigment, so the grains are dark red. In contrast, aabbcc produces no pigment, so the grains are white. See the following cross.

When dark red wheat (AABBCC) is crossed with white wheat (aabbcc), the F1 generation (AaBbCc) shows an intermediate red colour. In the F2 generation, during crossbreeding, a wide range of colours appears. Only one plant will darkest red grains (AABBCC) and one will plant will have white grains. While 62 plants will show different shades of red (moderate red to very light red).

For Information
Environmental factors like light, water and nutrients also influence the grain colour.

18.5.2- Epistasis

Most traits appear due to interactions between different genes. These interactions are of two main types.

1. In **allelic interactions**, alleles of the same gene interact with each other. Examples include incomplete dominance, codominance, and multiple alleles.

2. In **non-allelic interactions**, alleles of different genes (at different loci) interact with each other. A common example of this type is epistasis.

Epistasis is defined as the gene interaction, in which an allele masks or modifies the effect of the alleles of other genes (present at other loci). The allele which masks or modifies the effect is called **epistatic**. While, the allele whose effect is masked or modified is called **hypostatic**. For example, in humans, the allele of baldness is epistatic to the alleles of black and blonde hair.

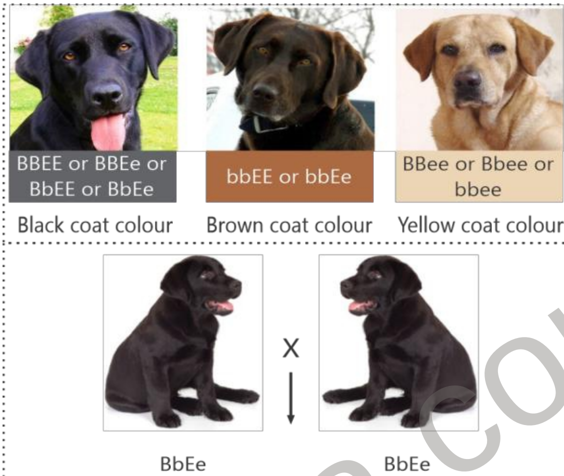
For Information
Both polygenic inheritance and epistasis involve more than one gene, but they work differently.
In polygenic inheritance, many genes act together, each having a small effect, to produce a trait. This results in continuous variation.
In epistasis, one allele masks or modifies the effect of another gene. This usually produces discontinuous variation (distinct categories of traits).

Coat Colour in Labrador

In many animals, coat colour is controlled by **epistasis**, where one gene affects another. For example, in Labrador dogs, coat colour (black, brown, yellow) depends on two genes.

1. The **B gene** controls pigment colour. Its dominant allele B gives black colour, while recessive b gives brown. So, BB or Bb will be black, and bb will be brown.
2. The **E gene** controls the deposition of pigment in fur. The dominant E allows pigment, but recessive e is epistatic to B or b. it blocks pigment deposition in fur. So, EE or Ee will pigment in fur, but ee will have yellow coat, no matter if B or b is present.

When two heterozygous black dogs (BbEe × BbEe) are crossed, the offspring show black, brown, and yellow colours in a 9:3:4 ratio.



Eggs \ Sperm	BE	Be	bE	be
BE	BBEE Black	BBEe Black	BbEE Black	BbEe Black
Be	BBEe Black	BBee Yellow	BbEe Black	Bbee Yellow
bE	BbEE Black	BbEe Black	bbEE Brown	bbEe Brown
be	BbEe Black	Bbee Yellow	bbEe Brown	bbee Yellow

Figure 18.8: Genotypes and phenotypes of coat colour in Labrador

Bombay Phenotype

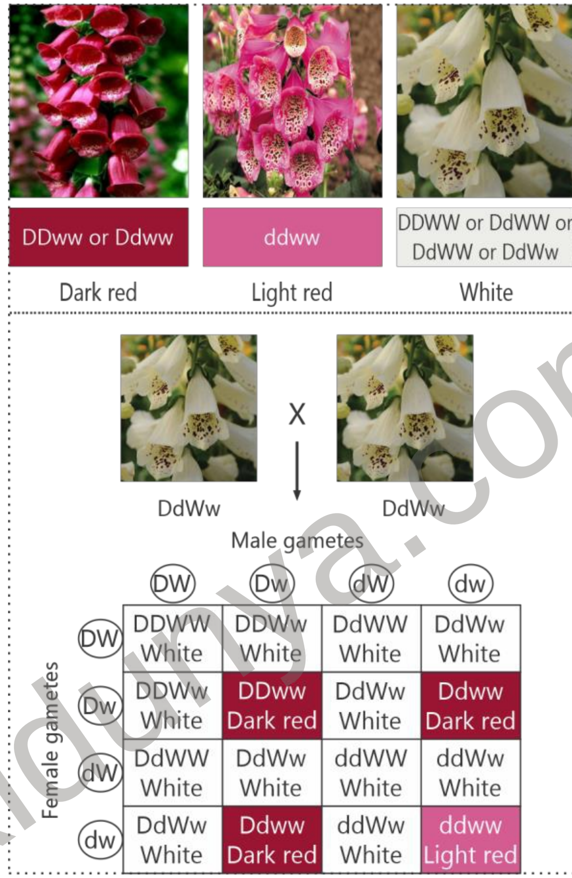
The expression of ABO genotypes also depends upon another gene i.e., H. It makes a particular H substance, which is a precursor to the A or B antigens. The alleles I^A and I^B produce enzymes which modify the H substance into antigen-A and antigen-B respectively. If an individual has I^A and I^B alleles but his genotype is hh, he will not produce H substance. So, his I^A and I^B alleles will not produce A and B antigens. In this way, he will have O phenotype due to the lack of A and B antigens. This phenotype is called Bombay phenotype because it was discovered in Bombay in 1952. It is mostly found in the India, Bangladesh, Pakistan, and Iran.

Flower Colour in Foxglove

In foxglove plants, the flower colour (dark red, light red, and white) is controlled by interaction in two genes (gene D and gene W).

1. Gene D has a dominant allele D that produces dark red petals. Its recessive allele d produces light red petals. Therefore, a genotype DD or Dd will have dark red and a genotype dd will have light red flower colour.
2. Gene W (present at another locus) has a dominant allele W that limits the distribution of pigment (produced by D and d) in the form of small spots so petals appear white from outside. The recessive w allows uniform pigment distribution.

The heterozygous plant DdWw will have white flowers (because the W allele limits the pigment in small spots in the inner side). If such heterozygous plant is self-fertilized, it produces plants with white, dark red and light red flowers in 12:3:1 ratio.



	DW	Dw	dW	dw
DW	DDWW White	DDWw White	DdWW White	DdWw White
Dw	DDWw White	DDww Dark red	DdWw White	Ddww Dark red
dW	DdWW White	DdWw White	ddWW White	ddWw White
dw	DdWw White	Ddww Dark red	ddWw White	ddww Light red

Figure 18.9: Genotypes and phenotypes of flower colour in foxglove

18.6- Gene Linkage and Crossing Over

In 1900, German geneticist **Karl Correns**, rediscovered Mendel's work and suggested the role of chromosomes in heredity. In 1902, American scientist **Walter Sutton** proposed the **chromosomal theory of inheritance**. This theory states:

- Genes are found at specific locations on chromosomes.
- The behaviour of chromosomes during meiosis explains Mendel's laws of inheritance.

18.6.1 Gene Linkage

In body cells, thousands of genes are present on a limited number of chromosomes. For example, humans have about 20,000 genes on 23 chromosome (present in two sets), and fruit fly (*Drosophila*) has about 13,000 genes on 4 chromosome (also present in two sets). This means many genes are arranged in a line on each chromosome, sometimes very close together. Genes present on the same chromosome are called **linked genes**, and this is known as gene linkage. Closely linked genes tend to stay together and do not assort independently during gamete formation. However, genes on different chromosomes, or those far apart on the same chromosome, assort independently.

Detection of gene linkage

Gene linkage and its frequency can be studied by performing crosses and observing how often different phenotypes appear in the offspring. The key idea is to compare parental combinations (same as parents) with recombinant or new combinations.

For Information
Gene linkage can only be observed when a cross produces large number of offspring. Larger the progeny size, the more likely it is to represent the actual frequency of linkage in the population.

- If offspring show parental and new phenotypes in equal 1:1:1:1 ratio, it means the genes are not linked and assort independently.
- If parental combinations are more frequent and new combinations are fewer, it indicates incomplete linkage, meaning genes are linked but can separate due to crossing over.
- If only parental combinations appear and no new combinations are seen, it shows complete linkage, meaning the genes are very close and do not separate.

Example of Detection of Linkage

American biologist, **Thomas Hunt Morgan,** studied two traits in *Drosophila* i.e., wing length and abdomen width. He observed that allele for long wings (L) is dominant over short or vestigial wing (l). Similarly, allele for broad abdomen (B) is dominant over narrow abdomen (b). Morgan did the following cross in drosophila;

Parents	Broad abdomen Long wings	X	Narrow abdomen Vestigial wings
		↓	
F1		Long wings Broad abdomens	

Then, he crossed two of F1 flies. The results are shown in the following figure:

Explanation of Result: According to law of independent assortment, the F2 generation should have the following 9:3:3:1 ratio:

F1 X F1	Long wings Broad abdomens	X	Long wings Broad abdomens
		↓	
F2	9 Long wings Broad abdomen (Parental combination)		1 Vestigial wing Narrow abdomen (Parental combination)
	3 Vestigial wings		3 Long-wings

	Broad abdomen (New combination)		Narrow abdomen (New combination)

But Morgan observed only parental combinations in F2. There were 3/4 flies with long wings-broad abdomen and 1/4 with vestigial wings-narrow abdomen.

F2	3 Long wings Broad abdomen (Parental combination)	1 Vestigial wing Narrow abdomen (Parental combination)

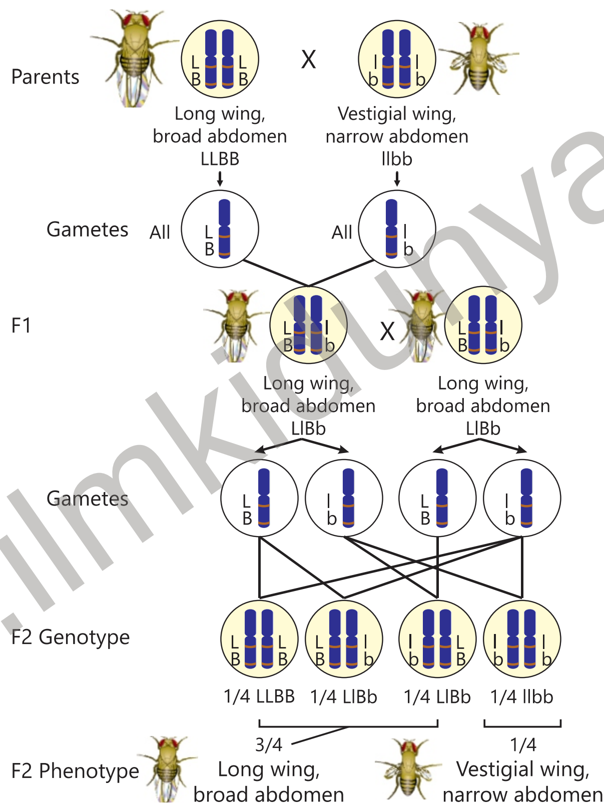


Figure 18.10: Gene linkage in Drosophila

From this data, Morgan concluded that the genes for wing length and abdomen width were located on the same chromosome and they did no assort independently.

18.6.2- Crossing Over

During the formation of gametes (meiosis-I), homologous chromosomes go through crossing over. This process is responsible for the recombination of linked genes. During crossing over, the chromatids of two homologous chromosomes line up next to one another. They swap (exchange) sections of DNA. In this process, DNA strands actually break and re-join. After crossing over, the homologous chromosomes separate from each other. The chromatids involved in crossing over still have the same genes in the same order, but the alleles have been rearranged. When these recombinant chromatids are distributed in different gametes, a wide variety is produced in gametes. Therefore, crossing over leads to recombination of genes and thus responsible for genetic variability.

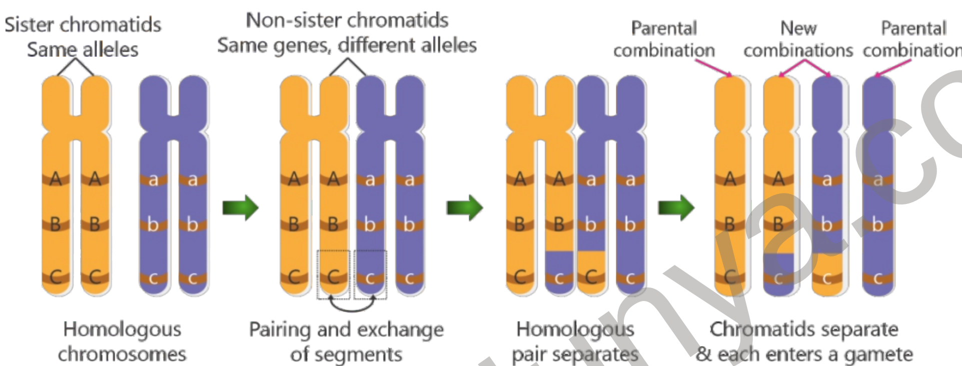


Figure 18.11: Crossing over

18.7- Sex Determination

The sex of an animal is determined during its embryonic development. In most animals, sex is determined on the basis of karyotype i.e., appearance of the complete set of chromosomes in the cells. Karyotype includes the numbers, sizes, and shapes of chromosomes. In the karyotype of most animals, all pairs of chromosomes are similar in female and male animals, except one pair. The chromosomes which are perfectly similar in female and male, are called **autosomes**. One pair of chromosomes is different in female and male and it determines the gender. The chromosomes of this pair are called **sex chromosomes**. The following are the main sex-determination systems on the basis of karyotype.

1- XX-XY System of Sex Determination

In *Drosophila*, mammals and most other vertebrates, the sex chromosomes pair consist of a rod-shaped **X chromosome** and a hook-shaped **Y chromosome**. Females have XX chromosomes while males have XY. Drosophila has 3 pairs of autosomes and 1 pair of sex chromosomes. Humans have 22 pairs of autosomes and 1 pair of sex chromosomes.

In these animals, females are **homogametic** which means that they produce all eggs with one X chromosome. On the other hand, males are **heterogametic** which means that they produce two types of sperm. Half carry one X chromosome and half carry one Y chromosome. If a sperm with X chromosome fertilizes the egg, the zygote will have XX chromosomes. This embryo will develop female sex organs and the baby will be a female. If a sperm with Y chromosome fertilizes the egg, the zygote will have XY chromosomes. This embryo will develop male sex organs and the baby will be a male.

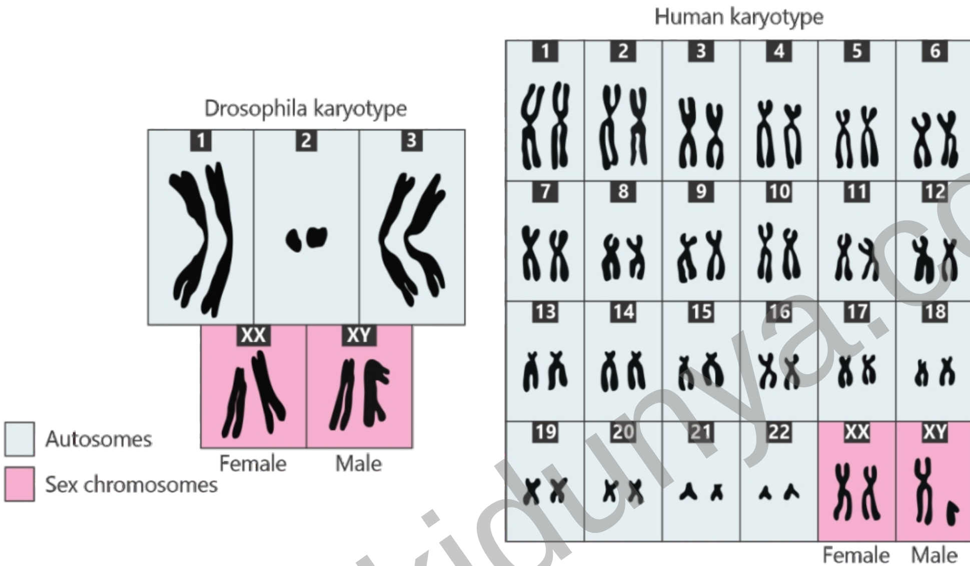


Figure 18.12: Autosomes and sex chromosomes in Drosophila and humans

2- XX–XO System of Sex Determination

This sex determination is found in some insects. For example, grasshopper has 22 autosomes (11 pairs). Male has total 23 chromosomes because its sex chromosome pair is XO i.e., only X chromosome is present and there is no Y chromosome. Male forms two types of sperm; half containing X and half without any sex chromosome. A gamete without any sex chromosome is called **nullo gamete**.

Female has 24 chromosomes. Its sex chromosome pair is XX. All eggs carry one X chromosome. If an X-carrying sperm fertilizes the egg, an XX female offspring is produced. If the nullo sperm fertilizes the egg, an XO male offspring is produced.

3- ZW–ZZ System of Sex Determination

This sex determination is common in birds. The autosome number differs in different birds but the sex chromosomes are ZW in females and ZZ in males. It means that the female bird produces two kinds of eggs; half with Z chromosome and half with W chromosome. While, the all sperms of the male have Z chromosome. When an Z carrying egg is fertilized by the sperm, offspring is male. When a Y carrying egg is fertilized by the sperm, offspring is female.

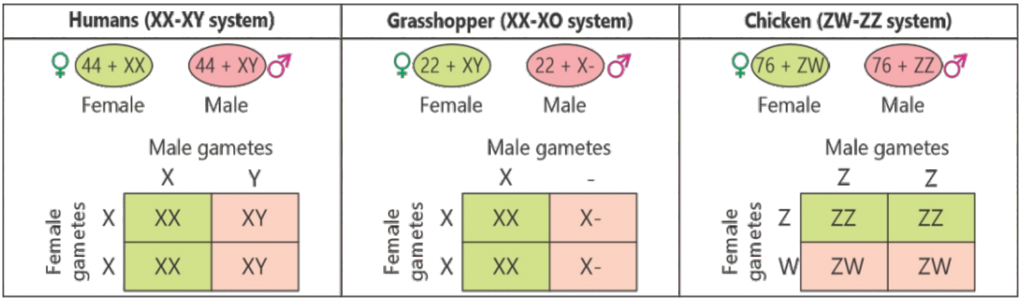


Figure 18.13: Sex determination in animals

18.8- SEX LINKAGE

The traits which are controlled by alleles present on sex chromosomes are called **sex-linked traits** and the phenomenon is called sex linkage. The sex-liked trait is actually **X-linked** because usually the Y chromosome carries almost no functional genes. That's why, the ratio of sex-linked traits is different in males and females. However, the alleles of very few traits are found on both X and Y. Such traits are equally distributed among males and females and are called **X-Y-linked** or **pseudo-autosomal** traits.

Discovery of Sex-Linkage

In 1910, Thomas Hunt Morgan observed a male *Drosophila* fly that differed from normal flies. This mutant male fly had white eyes instead of normal red. He crossed the mutant male to a normal (wild-type) female. All of the F1 progeny had red eyes.

Parents	1 White-eyed (male)	n	X	1 Red-eyed (female)
			↓	
F1	All Red eyed (male)			All Red-eyed (female)

He concluded that red eye colour was dominant over white. He crossed the red-eyed flies from the F1 generation with each other. Out of 4252 flies in F2, there were 782 (18%) white eyed. Although the ratio of red eyes to white eyes in the F2 was greater than 3:1, there was something important. All of the white-eyed F2 flies were males.

F1 X F1	1 Red-eyed (male)	X	1 Red-eyed (female)
		↓	
F2	3473 Red eyed (male & female)		782 White-eyed (male)

Morgan thought that perhaps it was impossible for a white-eyed female fly to exist. To test this idea, he test-crossed the F1 red-eyed female with the white-eyed male. He obtained both white-eyed and red-eyed males and females in a 1:1:1:1 ratio. Hence, it was proved that a female could have white eyes. Why, then, were there no white-eyed females among the progeny of the first cross? He mated a white-eyed female with a red-eyed male. All female offspring had red eyes, and all male offspring had white eyes.

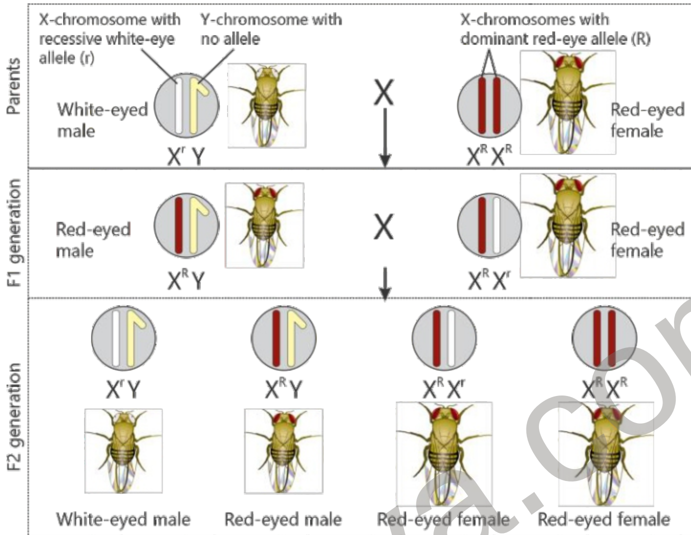


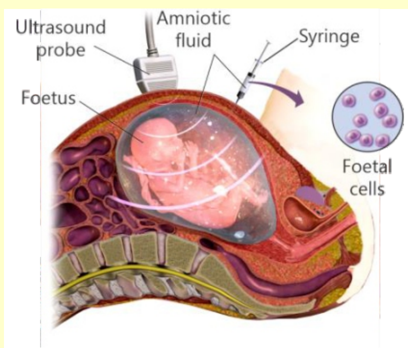
Figure 18.14: Inheritance of sex-linked trait (eye colour) in Drosophila

Morgan concluded that the gene of white-eye is present on X chromosome. He assumed that the Y chromosome did not have the allele of this gene. He called such genes as sex-linked genes and traits as sex-linked traits.

Embryonic Screening

Embryonic screening are the methods to check genetic disorders and abnormal number of chromosomes in embryos. It includes methods like ultrasound scan, chorionic villus sampling, and amniocentesis.

Amniocentesis is generally done during 15th and 20th week of pregnancy. In its procedure, a long needle is inserted into the amniotic sac. During it, an ultrasound probe locates the foetus and tests any abnormality. The inserted needle sucks some amniotic fluid which contains cells of the foetus. The foetal cells are kept in culture medium and checked for abnormal number of chromosomes.



Amniocentesis

18.8.1- Sex–linked Traits in Humans

X-linked Recessive Traits

The X-linked recessive traits are less common in females than males. It is because, such trait will appear in a female only when she has two recessive alleles on her two X chromosomes. While in males, this X-linked recessive trait will appear when only one allele is present on X-chromosome (because the Y-chromosome does not carry its allele).

So, this trait will appear in all the males who possess its allele. Examples of X-linked recessive traits include muscular dystrophy (muscle weakness and degeneration), red-green colour blindness and haemophilia (deficiency of clotting factors in the blood).

If a mother has the trait, all of her sons will also have it. If mother is a carrier (heterozygous) of the trait, 50% of her sons can inherit it. If such trait is present in father, all of his daughters will be carriers (daughters possess their father's X-chromosome) and no son will inherit this trait (sons do not inherit their father's X-chromosome).

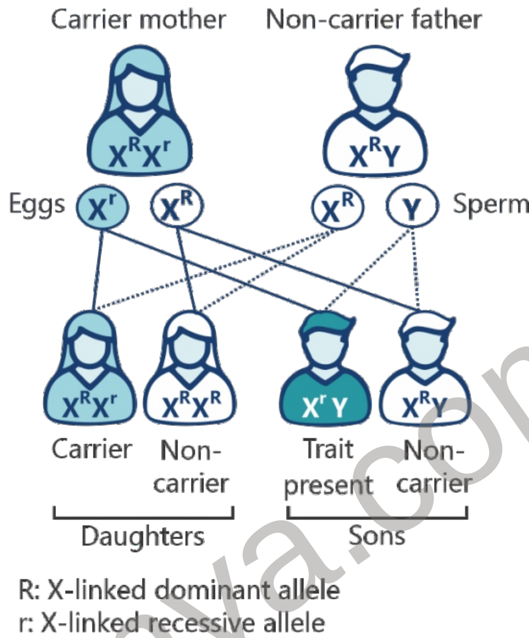


Figure 18.15: Inheritance of X-linked recessive traits

X-linked Dominant Traits

Such traits are in equal ratio in females and males. Examples include vitamin D-resistant rickets (inability to properly metabolize vitamin D, leading to softening and weakening of bones), Rett syndrome (neuro-developmental disorder with loss of speech, repetitive hand movements, and problems with walking), Aicardi syndrome (abnormal development of brain).

If an X-linked dominant trait is present in mother (homozygous or heterozygous), 50% of her offspring can inherit the trait. If such trait is present in father, all of his daughters will inherit it (a daughter inherits her father's X-chromosome) and no son will inherit it (a

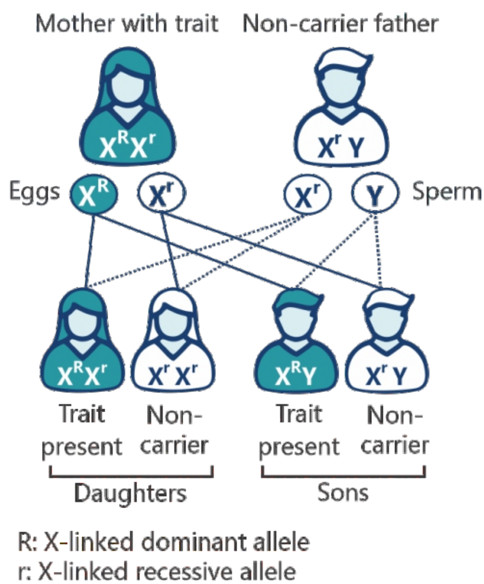


Figure 18.16: Inheritance of X-linked dominant traits

son does not inherit his father's X-chromosome).

Y-linked Traits

Y-chromosome contains relatively few genes compared to X chromosome. That's why Y-linked traits are relatively rare. The Y-linked genes do not have alleles on X chromosome. Such genes are called holandric genes (or hemizygous genes). Such genes are located only on the non-homologous region of Y-chromosome. The Y-linked traits appear only in males and are inherited from father to son only. The examples of Y-linked traits in human are hypertrichosis (growth of hair on the rim of pinna), porcupine man (straight hair on the body) and webbing of toes.

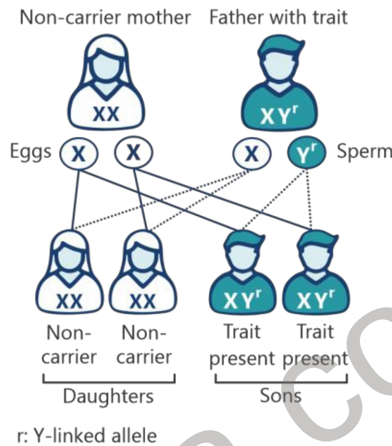


Figure 18.17: Inheritance of Y-linked traits

18.8.2- Sex Linked Disorders in Humans

1- Haemophilia

Haemophilia is a rare X–linked recessive trait. In this disease, the blood fails to clot properly after an injury. It is due to the reduction, malfunctioning, or absence of blood clotting factors. Haemophilia is of three types i.e., A, B and C.

Haemophilia A and B are due to abnormal blood clotting factors VIII and IX, respectively. Both these types are X–linked recessive traits. So, these are more common in males than females.

Haemophilia C is due to abnormal blood clotting factor XI. It is an autosomal recessive trait, so present in both genders equally.

For Information
In human males, there is a specific gene on Y-chromosome. It is known as SRY (Sex-determining Region Y). It produces a protein which starts development of testes and prevents the development of female reproductive structures.

Solve the following genetic problem.
If two normal parents have a haemophiliac child, what could be the genotypes of the parents?

Mechanism of Normal Blood Clotting (Coagulation)
Blood clotting is a complex process that helps to prevent excessive bleeding when an injury occurs. The mechanism involves the following steps:

1. When an injury occurs, platelets adhere to the site of injury, forming a platelet plug.
2. Activated platelets as well as the damaged tissue release of clotting factors.
3. The clotting factors perform chemical reactions (clotting cascade), in which soluble protein fibrinogen is converted into an insoluble protein called fibrin.

4. Fibrin forms a mesh-like structure that entraps parts of the blood and makes a solid clot.
5. The clot is stabilized by the cross-linking of fibrin strands.

If the balance of clotting system is disrupted, it can lead to abnormal clotting, known as **thrombosis**, which can cause serious health problems such as stroke and heart attack.

2- Colour-Blindness

In people with normal vision (**trichromatic** vision), there are three kinds of cone cells in the retina of eye – each sensitive for a specific primary colour. These are red-sensitive cones, green-sensitive cones and blue-sensitive cones. Each type contains its specific pigment proteins called **opsins**. The genes for red and green opsins are on X chromosome, while the gene for blue opsin is on autosome 7. The recessive alleles for opsins cause colour-blindness. It may be of following types.

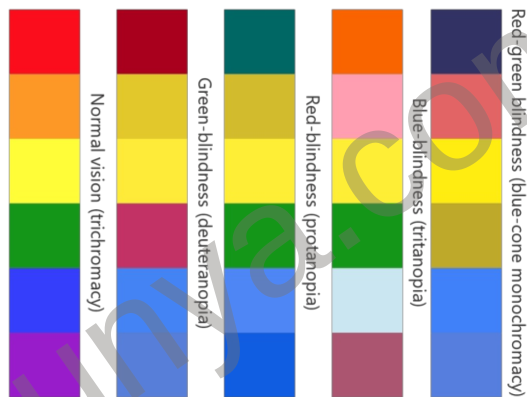


Figure 18.18: Dichromacy & monochromacy

Dichromacy: It is the perception of two primary colours and blindness of the third colour whose opsin is missing. It may be red-blindness (protanopia), green blindness (deuteranopia), or blue-blindness (tritanopia).

Monochromacy (total colour blindness)**:** It is the perception of a single colour and blindness of any two colours whose opsins are missing. In blue-cone monochromacy, red and green opsins are absent. It is also called red–green colour-blindness. Red-cone monochromacy and green-cone monochromacy are very rare.

There is no cure for colour blindness. However, there are assistive technologies such as colour-blind glasses and special software that can help individuals to differentiate between colours. Additionally, some colour-blind people have found success with colour discrimination training.

3- Muscular Dystrophy

There are more than 30 disorders classified as muscular dystrophies. **Duchenne muscular dystrophy** is the most common. It is X-linked recessive (so more common in males). About two thirds cases are inherited from a person's mother, while one third cases are due to a new mutation. If a male receives its recessive allele, he cannot make muscle proteins (dystrophin). It results in the death of muscle fibres and there is weakness and breakdown of skeletal muscles, especially those of the hips, pelvic area, thighs, and calves. It eventually progresses to the shoulders and neck, arms, respiratory and cardiac muscles, and other areas.

Muscular dystrophy affects a child around the age of four. It becomes evident when the child begins walking. By age 10, the child may need braces. By age 12, most patients are unable to walk and need wheelchair. Typical lifespans range from 15 to 45.

There is no cure for muscular dystrophy, but treatments are available to manage its symptoms and improve quality of life. For example; physical therapy is done to maintain strength, flexibility, and mobility; corticosteroid medicines can give short-term improvements in muscle strength and function; pain-killer medicines are used to relieve muscle pain; and surgery is done to correct joint contractures.

Figure 18.19: Symptoms of muscular dystrophy

18.8.3- Sex-limited and Sex-influenced Traits

Some traits are not sex-linked (because their alleles are not present on sex chromosomes) but there ratio is different in males and females. There are two categories of such traits.

1- Sex-limited Traits

The genes of such traits are present in both sexes but are expressed in only one sex due to anatomical or hormonal differences. Such traits are called sex-limited traits. For example, both male and female sheep have the genes of horn development. These genes are only expressed in male sheep. Similarly, the genes of milk production are present in both male and female mammals. But, these genes express only in female mammals. The genes of the growth of beard in humans are expressed in males only. Similarly, the genes of prostate cancer are expressed in males only and genes of ovarian cancer are expressed in females only, although both these genes are present in males and females.

For Information

Sex-limited genes are responsible for sexual dimorphism i.e., phenotypic difference between males and females of the same species regardless of genotype.

2- Sex-influenced Traits

In some traits, the same gene shows different expression in each sex. Sex-influenced genes have alleles which are dominant in one sex and recessive in the other. This difference in expression is due to hormonal difference between the males and females. For example, in humans, the traits of the amounts of body hair and baldness are sex-influenced traits. The allele of baldness is dominant in men but recessive in women. So, a man can be bald when he is homozygous (BB) or heterozygous (Bb) for baldness alleles. While a woman can be bald only when she inherits two recessive alleles (bb).

For Information

Genetics has made significant progress. Now, it is more applied and practical science. For example:

- DNA sequencing and genetic engineering, have enabled researchers to manipulate genes.
- The completion of Human Genome Project has led to a deeper understanding of the genetic basis of various diseases and traits.
- Genetics plays important role in healthcare, helping doctors to diagnose and treat patients based on their unique genetic profiles.

EXERCISE

SECTION 1: MULTIPLE CHOICE QUESTIONS

1. In the monohybrid cross of Mendel, when a pure round-seeded plant was crossed with a winkled-seeded plant, all the F1 progeny were round-seeded, because of:
   (a) Co-dominance (b) Incomplete dominance
   (c) Complete dominance (d) Epistasis
2. In a cross of AaBb x AaBb, what fraction of the offspring can be expected to express one of the two dominant alleles, but not both?
   (a) 1/16 (b) 6/16 (c) 9/1 (d) 16/16
3. In which of these cases, two genes can experience independent assortment?
   (a) They are located in close proximity on the same chromosome.
   (b) Crossing over between the genes does not occur.
   (c) They are located on different chromosomes or are far apart on the same chromosome.
   (d) The expression of one gene does not affect the expression of the other.
4. The blood group AB may be given to a person who has blood group;
   (a) A (b) O (c) B (d) AB
5. If father has blood group B and the mother has blood group A, what blood groups their children have?
   (a) A or B only (b) AB only (c) AB or O only (d) A, B, AB or O

6. A gene for corn has two alleles, one for yellow kernels and one for white kernels. Cross pollination of yellow corn and white corn results in ears with approximately equal number of yellow and white kernels. Which term best describes the relationship between the two alleles?
   (a) Crossing over (b) Recombination
   (c) Incomplete dominance (d) Codominance
7. A cross between two plants with genotypes AaBb and AaBb results in an F1 generation that is 25% AABB, 50% AaBb, and 25% aabb. Which reason most likely explains why other possible genotypes are not present?
   (a) The genes underwent independent assortment
   (b) The loci of the genes are close together.
   (c) The loci of the genes are on different chromosomes.
   (d) Crossing over occurred between chromosomes.
8. In pea plants, the allele for tall stalks (T) is dominant over the allele for short stalks (t). Suppose a cross between a tall pea plant and a short pea plant produces 43 tall offspring and 47 short offspring. If one of the tall offspring is crossed with one of the short offspring, what ratio of genotypes would be most likely in their offspring?
   (a) 1 TT : 2 Tt : 1 tt (b) 3 Tt : 1 tt
   (c) All Tt (d) 1 Tt : 1 tt
9. Allele B produces black coat colour while b produces white coat colour. Allele C produces curly fur while c produces straight fur. In a cross between two BbCc parents, predict the fraction of offspring with black coat colour and straight fur.
   (a) 1/16 (b) 9/16 (c) 3/4 (d) 16/16
10. What is the criterion to mark an allele as dominant?
   (a) The frequency of the allele in population
   (b) The expression of the allele in heterozygous genotype
   (c) The location of the allele on chromosome
   (d) The frequency of the crossing over of the allele
11. Which of these characters is controlled by a multiple allelic system?
   (a) ABO blood group system (b) Rh blood group system
   (c) MNS blood group system (d) All of these
12. Albinism is caused by a recessive gene. A woman with albino father marries an albino man. What will be the proportion of her progeny?
   (a) 2 normal : 1 albino (b) All normal
   (c) All albino (d) 1 normal : 1 albino
13. When the activity of one gene is suppressed by the activity of a non-allelic gene, it is known as;
   (a) Incomplete dominance (b) Epistasis
   (c) Complete dominance (d) Multiple allelic effect

14. Which of the following is a result of Erythroblastosis fetalis?
(a) Anaemia in the foetus
(b) Jaundice in the foetus
(c) Mental retardation in the foetus
(d) All of these

15. Colour blindness is more common in males because it is a/an;
(a) Autosomal trait
(b) X-linked dominant trait
(c) X-linked recessive trait
(d) Y-linked trait

SECTION 2: SHORT QUESTIONS

1. Name the main exceptions to the Mendel's laws of inheritance.
2. What is probability? How does probability relate to genetics?
3. Justify that multiple alleles provide many different phenotypes.
4. Name various human blood group systems.
5. What do you mean by multiple alleles? Name the alleles responsible for ABO blood group system.
6. What is co-dominance. Give an example.
7. What antigens and antibodies are found in ABO blood group system?
8. Justify why O-negative individuals are called universal donors and AB-positive individuals are called universal recipients.
9. Why is human male referred as heterogametic?
10. What is gene linkage?
11. How can the linked gene assort independently?
12. List at least five polygenic traits in humans.
13. Write brief note on Y-linked inheritance in humans.
14. Define sex linkage and give an example.
15. Solve the following genetic problems:
    i. A plant of genotype AABbCC is self-pollinated. Calculate the phenotypic ratio of F2 generation.
    ii. In four-o'clock plant, the red and white flower colours show incomplete dominance. Calculate the ratios in crosses between (i) two red-flowered plants; (ii) a red-flowered and a pink-flowered plant; (iii) a pink-flowered and a white-flowered.
    iii. A man with blood group A marries a woman who has blood group AB. What blood groups are expected among their children?
    iv. Two white sheep produce a black offspring. What are the parents' genotypes for the colour? What is the probability that their next offspring will be black?
    v. Albinism (lack of pigment) in man is caused by a recessive gene. If normal parents have an albino child, what is the probability that their next child will be normal for colour?
16. Differentiate between:
    - Gene and allele
    - Genotype and phenotype

- Homozygous and heterozygous
- Monohybrid and dihybrid cross
- Incomplete dominance and co-dominance
- Epistasis and polygenic inheritance
- XX-XY and ZZ-ZW sex determination
- XX-XY and XX-ZO sex determination
- Sex-linked and sex-limited traits
- Sex-limited and sex-influenced traits

SECTION 3: LONG QUESTIONS

1. Explain Mendel's experiment which led to the formulation of the law independent assortment.
2. Explain incomplete dominance and exemplify it through the inheritance of flower colour in 4 O' clock plant.
3. Explain Erythroblastosis foetalis in the light of antigen-antibody reaction. What measures can be taken to prevent Erythroblastosis foetalis?
4. Describe polygenic inheritance, using an example from grain colour in wheat.
5. Explain epistasis with the help of an example.
6. Explain how gene linkage counters independent assortment and crossing-over modifies the progeny.
7. Exemplify the concept of gene linkage by quoting the example of wing length and width of abdomen in Drosophila melanogaster.
8. Explain the XX-XY mechanism of sex determination in Drosophila and mammals.
9. Describe the XX-XO and ZZ-ZW sex determination systems and evaluate by studying the karyotype.
10. Explain the inheritance of a sex-linked trait in Drosophila.
11. Describe sex-influenced and sex-limited traits with common examples from human genetics.
12. Write a note on the inheritance of haemophilia in humans.
13. Write a note on the inheritance of colour blindness in humans.
14. Explain Morgan's experiment to explain crossing over and gene linkage.

INQUISITIVE QUESTIONS

1. Collect data from the class or the college to see how many individuals have AB blood group and construct a pie chart and histogram for the collected data.
2. Use a dice to calculate how many times out of 100 throws can you get sixes.
3. Derive an idea to get alternatives of blood transfusion. (reference could be made to synthesized plasma and serum).
4. Justify why a recessive blood group allele of 'i' is more frequent in population.
5. Justify blood donation as a service to suffering humanity.
6. Suggest ways to save lives through the knowledge gained in this chapter.