CHAPTER 12

# INHERITANCE

## TERMINOLOGY

1. **Genes/Factors**
   - **Definition**: Units of heredity that are made up of DNA and determine specific characteristics or traits.
   - **Example**: The gene for eye color in humans.
2. **Locus**
   - **Definition**: The specific physical location of a gene or other significant sequence on a chromosome.
   - **Example**: The locus of the gene for the hemoglobin beta chain is on the short arm of chromosome 11.
3. **Trait**
   - **Definition**: A specific characteristic or feature exhibited by an organism, which can be inherited, and at least have two alternatives.
   - **Example**: Hair color in humans is a trait that can vary from blonde to black.
4. **Alleles**
   - **Definition**: Different versions or forms of a gene that can exist at a specific locus.
   - **Example**: The ABO blood type gene has three alleles: A, B, and O.
5. **Homozygous Alleles**
   - **Definition**: Having two identical alleles for a particular gene at the same locus.
   - **Example**: An individual with two A alleles (AA) for blood type.
6. **Heterozygous Alleles**
   - **Definition**: Having two different alleles for a particular gene at the same locus.
   - **Example**: An individual with one T allele and one t allele (Tt) for tallness.
7. **Dominance**
   - **Definition**: A relationship between alleles of a gene, in which one allele masks the expression of another allele.
   - **Example**: In pea plants, the allele for purple flowers (P) is dominant over the allele for white flowers (p).
8. **Recessive**
   - **Definition**: An allele whose effects are masked in the presence of a dominant allele.
   - **Example**: The allele for white flowers (p) in pea plants is recessive to the allele for purple flowers (P).
9. **Phenotype**
   - **Definition**: The observable physical or biochemical characteristics of an organism, resulting from the interaction of its genotype with the environment.
   - **Example**: A person's blood type, such as A, B, AB, or O.
10. **Genotype**
    - **Definition**: The genetic makeup of an organism, consisting of both the visible and non-visible alleles.
    - **Example**: The genotype for blood type could be AA, AO, BB, BO, AB, or OO.
11. **Gene pool**
    - **Definition**: The complete set of different alleles in a population.
    - **Example**: The diverse set of alleles for coat color in a population of wild rabbits.
12. **Monohybrid Cross**
    - **Definition**: A genetic cross between two individuals that differ in one trait.
    - **Example**: Crossing a pea plant with yellow seeds (YY) with one that has green seeds (yy).

11. **Transcription takes place in:**
A. Cytoplasm B. Nucleus
C. Ribosomes D. Endoplasmic reticulum

12. **Why are variations important in biology?**
A. They maintain genetic similarity
B. They contribute to species adaptation
C. They reduce genetic diversity
D. They stabilize genetic expression

**Answer Key**

| 1. B | 2. C | 3. B | 4. C | 5. C | 6. B | 7. B | 8. C | 9. C | 10. B | 11. B | 12. B |
|---|---|---|---|---|---|---|---|---|---|---|---|



**SLO 12.2 — Mendelian Inheritance**

**Students will be able to:**

**12.2 Explain the law of segregation and independent assortment using a suitable example related to the pea plants.**

# MENDELIAN INHERITANCE

- Genetics, as a science, began in 1900 after the rediscovery of an article first published in 1866. The article was written by Gregor Johann Mendel.

### Contributions of Gregor Johann Mendel

- Gregor Johann Mendel was an Augustinian monk. He was the pioneer in explaining the mechanisms of inheritance.
- His groundbreaking research involved experimenting with pea plants to understand **how traits are passed** from one generation to the next.

## Association of Inheritance with Mendel's Laws

### Development of Pure Breeding Plants

- In the 1860s, Gregor Mendel started his experimental work by developing **true** or **pure-breed** varieties of pea plants.
- A pure breeding plant is defined as a plant that consistently produces offspring identical to itself when self-fertilized.
  - For example, self-fertilizing a round seed-shaped plant that results in offspring with round seeds is a pure breeding plant.
- To establish a pure breeding plant, Mendel repeatedly self-fertilized pea plants over several generations until a consistent **phenotype** was achieved.

### Hybridization Techniques

- Mendel's second step involved hybridization. Hybridization is the cross-fertilization between two plants with different traits.
  - A **monohybrid** cross involves the breeding of two plants that **differ in a single trait**, such as crossing a plant with round seeds with another plant that has wrinkled seeds.
  - A **dihybrid** cross involves breeding two plants that **differ in two distinct traits**, such as crossing a plant with round, yellow seeds with another plant that has wrinkled, green seeds.

### Mendel's Experimental Approach

- Mendel's successful approach included the study of single traits first and then the analysis of combinations of two or three traits.
- This methodical approach allowed him to observe how traits were inherited independently or together.

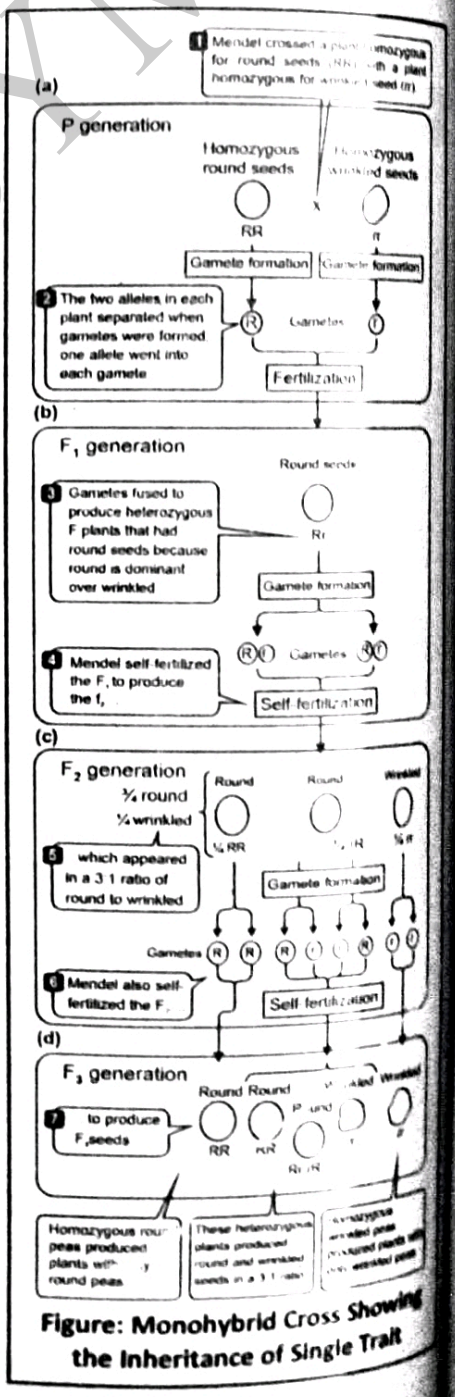


**Figure: Monohybrid Cross Showing the Inheritance of Single Trait**

- Based on his detailed observations, Mendel formulated two foundational generalizations. These are known as Mendel's laws of inheritance.
- These laws explain how traits are transmitted from parents to offspring.

## Inheritance of a Single Trait (Monohybrid Cross)

### Overview of Monohybrid Cross

- Mendel's studies on the inheritance of single traits involved the **monohybrid cross**, where two plants differing in only one trait are crossed.

### Experimental Procedure and Initial Observations

- Mendel crossed a pure breeding plant with **round seeds** with another pure breeding plant that produced **wrinkled seeds**.
- In these experiments, the first generation of offspring (F1) uniformly exhibited the round seed phenotype of one of the parents.

### Generational Observations

- When the F1 generation plants were self-fertilized, the resulting second generation (F2) displayed a 3:1 ratio of round to wrinkled seed shapes.
- Mendel consistently observed this 3:1 ratio in all seven contrasting pairs of traits he studied.

### Further Generational Study

- Mendel continued his experiments by self-fertilizing the F2 generation to produce a third generation (F3).
- He noted that **one-third** of the F2 plants with round seeds **bred true** to produce only round seeds, suggesting they were pure breeding like the original parent (P1).
- Two-thirds of the F2 plants with round seeds produced both round and wrinkled seeds in a 3:1 ratio, similar to the F1 round seeds, indicating they were not pure breeding.
- All F2 plants with wrinkled seeds produced only wrinkled seeds, indicating they were pure breeding like their respective P1 ancestor.

## Interpretations of Mendel's Results

### Introduction to Genetic Factors



- Mendel introduced the concept that contrasting forms of a trait, such as seed shape in peas, are determined by what he called "elementens." The elementens are now known as genes.
- Each pea plant possesses a pair of genes. These are referred to as alleles. These alleles occupy the same position (locus) on chromosomes.
- These alleles, one inherited from each parent, combine to form the plant's genotype. This **genotype** determines the **observable characteristics** or **phenotype** of the plant.

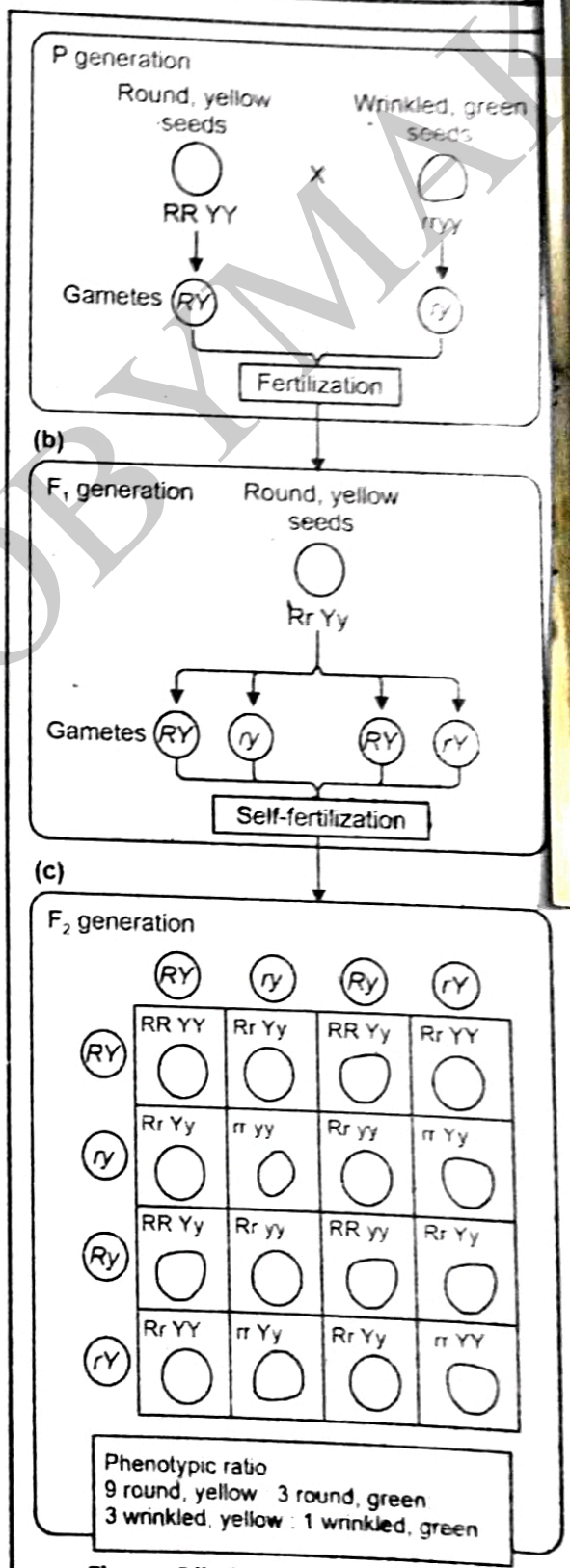


**Figure: Dihybrid Cross Showing the Inheritance of Two Traits**

### Dominance and Recessiveness

- Mendel identified alleles that mask the expression of others as "**dominant**," and those that are masked as "**recessive**."

- In his studies, he used **uppercase letters to represent dominant alleles** (e.g., "R" for round seeds) and **lowercase letters for recessive alleles** (e.g., "r" for wrinkled seeds).

### Homozygous and Heterozygous Conditions

- An organism with two identical alleles for a trait is described as **homozygous**, and this individual is called a **homozygote**.
- True-breeding plants from Mendel's P1 generation showed homozygous genotypes, "RR" for round seeds and "rr" for wrinkled seeds.

### Mendel's Laws of Segregation

- According to law of segregation, "the two coexisting alleles for each trait in an individual segregate (separate) from each other at meiosis, so that each gamete receives only one of the two alleles".
- Mendel concluded that alleles separate during gamete formation, resulting in each gamete carrying only one allele for each trait.
- Fertilization is a random process, restoring the pair of alleles in the offspring. For example, the union of "R" from one gamete and "r" from another would result in the "Rr" genotype in the offspring.
- The offspring, therefore, can exhibit **heterozygosity**, having two different alleles for a trait. It is referred to as a heterozygote.

### Genetic Ratios and Punnett Square

- The use of a Punnett square helps predict that among the F2 progeny, there will be a distribution of "RR" (homozygous round), "Rr" (heterozygous round), and "rr" (homozygous wrinkled) in a typical 1:2:1 genotypic ratio.
- Mendel observed a 3:1 phenotypic ratio in the F2 generation, which matched the predicted outcomes based on allele segregation.
- This consistent observation across seven different traits led Mendel to formulate the Law of Segregation, stating that each gamete receives only one allele of a gene pair, which then randomly unite during fertilization to form the zygote.
- Mendel's work laid the foundational principles of modern genetics. It showed how traits are inherited through discrete units (later called genes) and how these units exhibit dominance, segregation, and independent assortment.

## Inheritance of Two Traits (Dihybrid Cross)

- Dihybrid cross is a cross between two individuals that are **different in two traits.**
- The inheritance of two traits simultaneously can be studied in a dihybrid cross.

### Mendel's Study on Seed Colour and Shape

- Two of the seven characters Mendel studied were seed **colour** and **shape**.
- The shape of the seeds may be either round (**dominant**) or wrinkled (**recessive**), and the colour of the seeds may be either yellow (**dominant**) or green (**recessive**).



### Procedure and Observations

- When Mendel crossed a homozygous round yellow (RRYY) plant with a homozygous wrinkled green (rryy) plant, all the offspring in the F1 generation exhibited both dominant phenotypes, i.e., round yellow.
- To analyze the genotype of the F1 plants, he self-fertilized them to produce the F2 generation. He expected that the dominant and recessive combinations would be produced in a 3:1 ratio in the F2 generation, as he had observed in the monohybrid cross.
- However, he observed that the offspring were produced in four phenotypic combinations: round yellow, round green, wrinkled yellow, and wrinkled green, in the ratio of 9:3:3:1.
- The occurrence of **recombinant phenotypes**, such as round green and wrinkled yellow, was surprising.

### Interpretations of the Results

- Based on these observations, Mendel concluded that the F1 offspring (round yellow) were dihybrid, i.e., heterozygous (RrYy) for both traits.

- The key event in the experiment occurred when the F1 plants self-pollinated and produced F2 offspring.
- An F1 plant can produce four classes of gametes (RY, rY, Ry, and ry) in equal quantities.
- If the sperms of the four classes are crossed with eggs of the four types, there will be 16 (4×4) equally probable ways in which the alleles can combine in the F2 generation, as shown in the Punnett square in Figure.
- These combinations result in four phenotypic categories with a ratio of 9:3:3:1.

### Law of Independent Assortment

- Mendel tested his seven pea characters in various dihybrid combinations and always observed a 9:3:3:1 phenotype ratio in the F2 generation.
- The results of Mendel's dihybrid experiments are the basis for what we now call the **law of independent assortment**, which states that "each pair of alleles assort independently of other pairs of alleles during gamete formation."
- In other words, the alleles of each pair of contrasting traits have an equal probability of assorting with the alleles of another pair.



**Test Your Skills**

1. Why do some historians and scientists consider Mendel "lucky" in his choice of pea plant traits?
2. How might Mendel's conclusions have differed if the traits he studied were linked on the same chromosome?
3. Provide examples of how Mendel's laws are applied in genetic counseling and disease prevention.
4. How did Mendel apply the principles of probability to predict the outcomes of genetic crosses?
5. In what ways did Mendel's work lay the groundwork for the discovery of DNA and the modern field of genetics?
6. What is the purpose of a test cross, and how did Mendel use it to determine the genotype of an organism?
7. How did Mendel use genetic ratios to predict the outcomes of his crosses?
8. How did Mendel's observations of the F2 generation lead to the formulation of the Law of Segregation?
9. How did Mendel's dihybrid crosses provide evidence for the Law of Independent Assortment?
10. Why was it important for Mendel to develop pure breeding lines before proceeding with his hybridization experiments?

## Multiple Choice Questions

### Mendelian Inheritance

1. **Who rediscovered the principles of genetics in 1900?**
   A. Charles Darwin
   B. Gregor Johann Mendel
   C. Thomas Hunt Morgan
   D. Francis Crick
2. **What organism did Gregor Mendel use for his experiments?**
   A. Fruit flies B. Mice
   C. Pea plants D. Yeast
3. **What is a pure breeding plant?**
   A. A plant with mixed traits
   B. A plant used in hybridization
   C. A plant that undergoes mutation
   D. A plant that produces offspring identical to itself when self-fertilized
4. **What does a monohybrid cross involve?**
   A. Crossing two different species
   B. Crossing two plants differing in one trait
   C. Crossing two plants with the same traits
   D. Crossing two genetically identical plants
5. **How did Mendel ensure his pea plants were pure breeding?**
   A. By hybridization
   B. By self-fertilizing them over several generations
   C. By using chemicals
   D. By genetic modification
6. **What phenotype did the F1 generation consistently exhibit in Mendel's experiments?**
   A. Wrinkled seeds B. Mixed traits
   C. Round seeds D. No seeds
7. **What ratio did Mendel observe in the F2 generation for round to wrinkled seeds?**
   A. 1:1 B. 3:1
   C. 2:1 D. 4:1

8. **What term did Mendel use to describe the units of inheritance?**
   A. Chromosomes B. Elementens
   C. Particles D. Cells
9. **What are alleles according to Mendel's research?**
   A. Types of proteins
   B. Forms of DNA
   C. Variants of a gene
   D. Types of chromosomes
10. **What describes an organism with two different alleles for a trait?**
   A. Homozygous B. Heterozygous
   C. Hybrid D. Mutant
11. **Which law states that each gamete receives only one allele of a gene pair?**
   A. Law of Independent Assortment
   B. Law of Dominance
   C. Law of Segregation D. Law of Uniformity
12. **How are Mendel's observations summarized in terms of genetic ratios for a monohybrid cross?**
   A. 1:1:1:1 B. 2:1:1
   C. 1:2:1 D. 3:3:1
13. **What does the 3:1 phenotypic ratio in the F2 generation suggest about the alleles?**
   A. They are codominant
   B. They are incomplete dominant
   C. One is dominant over the other
   D. They are both recessive
14. **How did Mendel use the concept of probability in genetics?**
   A. To predict weather patterns
   B. To calculate genetic drift
   C. To predict outcomes of genetic crosses
   D. To study population genetics

**Answer Key**

| 1. B | 2. C | 3. D | 4. B | 5. B | 6. C | 7. B | 8. B | 9. C | 10. B | 11. C | 12. C | 13. C | 14. C |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|

**SLO 12.3 to 12.6 — Law of Independent Assortment**

Students will be able to:

12.3 Relate the law of independent assortment to the random orientation of chromosomes during meiosis.
12.4 Express limitations of independent assortment and its usefulness.
12.5 Show that independent assortment leads to variation in the gametes.
12.6 Evaluate that the inheritance of genes and their mixing during fertilization is based on mathematical probabilities.



## LAW OF INDEPENDENT ASSORTMENT

### Overview of Meiosis

- According to the law of independent assortment, "each pair of alleles assort independently of other pairs of alleles during gamete formation."
- In other words, the alleles of each pair of contrasting trait have equal probability to assort with the alleles of other pair.

### Mechanism of Independent Assortment

- During metaphase I of meiosis, each homologous pair of chromosomes aligns independently at the cell's equator, which is crucial for independent assortment.
- The first division of meiosis results in the sorting of maternal and paternal homologues into daughter cells independently of other chromosome pairs.

### Genetic Variability

- The independent positioning and separation of chromosome pairs during metaphase I lead to a variety of genetic combinations.

**Science Titbits**

**a.** Observation, assumption, experimentation, and creativity, all of them are evident in Mendel's approach. The experiments performed by Mendel were elegant and his conclusions constitute the foundation of the modern science of Genetics. Mendel is therefore appropriately called, the father of Genetics.

**b.** R. C. Punnet devised what is known as the Punnett square for summarizing the fusion of gametes in genetic crosses. Punnett was a Professor of Genetics at Cambridge University. He wrote a large number of papers between 1900 and 1958, most of which helped to confirm and extend Mendel's work.

- Humans have 23 pairs of chromosomes. Thus, the number of possible combinations of maternal and paternal chromosomes in gametes is $2^{23}$, which is approximately equal to eight million different combinations.
- Each gamete produced during an individual's lifetime contains one of these eight million potential combinations of chromosomes inherited from both parents.

## Limitations of Mendel's Law of Independent Assortment

### Applicability Limitations

- Mendel's principles are the basis for understanding heredity. However, they do not apply to all biological situations.
- His work is relevant primarily to diploid organisms. Organisms that are not diploid do not conform to these principles.



### Genetic Linkage

- Genes located on the same chromosome typically do not assort independently.
- If an offspring inherits one trait from a chromosome, they are likely to inherit other linked traits unless crossing over occurs during meiosis.

### Sex-Linked Traits

- Genes on the X chromosome follow a unique pattern of inheritance.
- Males, having only one X chromosome and a Y chromosome with fewer genes, are more prone to express recessive X-linked traits.

### Significance of Independent Assortment

**Contribution to Genetic Variation**

- Independent assortment is a major mechanism that contributes to genetic diversity in successive generations.
- This process, along with mutations and crossing over, allows traits to combine in new ways in offspring.

**Role in Adaptation**

- The new combinations of traits resulting from independent assortment and crossing over can enhance an organism's ability to adapt to different environments.
- These variations are often crucial for survival and evolution, allowing species to adjust to new or changing environments.

## Usefulness and Scope of Independent Assortment

### Role in Genetic Variation

- Independent assortment is a crucial source of genetic variation, alongside mutation and crossing over.
- This process contributes to the diversity of traits across generations.

### Impact on Evolution and Adaptation

- Independent assortment, combined with crossing over, allows traits to recombine in new ways in subsequent generations.
- These new combinations of characteristics can provide necessary adaptations for survival in changing environments.

## Independent Assortment and Variation in Gametes

### Genetic Variation through Meiosis

- Meiosis produces genetically distinct gametes, each with unique combinations of chromosomes.
- Independent assortment during meiosis leads to these variations by randomly distributing maternal and paternal chromosomes into gametes.

### Mechanisms Contributing to Genetic Variation

- **Crossing Over**: Occurs in prophase I of meiosis, where sections of DNA are exchanged between homologous chromosomes, creating new genetic combinations.
- **Random Assortment of Chromosomes**: Takes place in metaphase I, where chromosomes are arranged randomly at the cell's equator before being separated.
- **Random Fusion of Gametes**: Involves the combination of egg and sperm from different parents, leading to the creation of unique zygotes.

### Significance in Offspring Diversity

- These mechanisms ensure high levels of genetic diversity among offspring, which is vital for adaptation and survival in varying environmental conditions.



## Inheritance and Mathematical Probabilities

### Basic Concept of Probability

- Probability is the likelihood that a particular event will occur.
- It is calculated as the number of favorable outcomes divided by the total number of possible outcomes ($P = a/n$). Here, 'P' is the probability, 'a' is the number of favorable outcomes, and 'n' is the total number of possible outcomes.

### Examples in Everyday Life

- For example, when we toss a coin, the probability (P) of getting a head or a tail is ½. That is, there is one favorable outcome out of two possible outcomes.

### Mendel's Application of Probability to Genetics

- Gregor Mendel, with his strong background in mathematics, applied the principles of probability to genetic inheritance.
- He realized that the segregation of alleles during gamete formation and their recombination at fertilization follow predictable probabilistic rules.

### Probability in Genetic Crosses

- In genetics, the probability of inheriting a specific phenotype can be calculated for different generations.
  - In a **monohybrid cross**, the probability of inheriting the dominant phenotype in the $F_1$ generation is 100% and the recessive phenotype is 0%.
  - In the $F_2$ generation, the probability of showing the dominant phenotype is 3/4, while that of the recessive phenotype is 1/4.

### Combining Probabilities of Independent Events

- The probability of two or more independent genetic events occurring together can be determined using the product rule, which multiplies the probabilities of the individual events.

**Test Your Skills**

1. Why are Mendel's principles primarily applicable to diploid organisms, and how do they fall short for non-diploid organisms?
2. How does crossing over during meiosis affect the inheritance of linked genes?
3. How does independent assortment contribute to genetic diversity in offspring?
4. How does the new combination of traits resulting from independent assortment enhance an organism's ability to adapt?
5. Why is high genetic diversity among offspring crucial for survival in different environmental conditions?
6. How can the probabilities of different genotypes and phenotypes in offspring be predicted using the rules of probability?
7. In a monohybrid cross, what are the probabilities of inheriting dominant and recessive phenotypes in the $F_1$ and $F_2$ generations?

## Solve at least four genetic problems, to illustrate the law of independent assortment.

**12.1** **PKU and albinism are two autosomal recessive disorders, unlinked in human beings. If a couple, each of them heterozygous for both traits, produce a child, what is the chance of their having a child with: (a) PKU? (b) albinism? (c) Both traits?**

**Sol.** To solve the genetics problem, we can use the basic principles of probability. Each parent is heterozygous for both PKU (Pp) and albinism (Aa).

**Step 1: Understand the Probability of Inheriting Each Trait**

Since both PKU and albinism are autosomal recessive traits, a child must inherit two recessive alleles (one from each parent) to express the disorder.

For a parent who is heterozygous (Pp or Aa), the probability of passing on the recessive allele (p or a) is 1/2, and the probability of passing on the dominant allele (P or A) is also 1/2.

**Step 2: Calculate the Probabilities for Each Trait**

**(a) Chance of having a child with PKU (pp):**

- Probability of inheriting one recessive allele (p) from one parent: 1/2
- Probability of inheriting one recessive allele (p) from the other parent: 1/2

To have PKU, the child must inherit the recessive allele from both parents:

Probability of pp = 1/2 × 1/2 = 1/4

Therefore, the chance of having a child with PKU (pp) is 1/4 or 25%.

**(b) Chance of having a child with albinism (aa):**

- Probability of inheriting one recessive allele (a) from one parent: 1/2
- Probability of inheriting one recessive allele (a) from the other parent: 1/2

To have albinism, the child must inherit the recessive allele from both parents:

Probability of aa = 1/2 × 1/2 = 1/4

Therefore, the chance of having a child with albinism (aa) is 1/4 or 25%.

**(c) Chance of having a child with both PKU and albinism (pp and aa):**

Since PKU and albinism are unlinked traits, the probability of having both traits is the product of the individual probabilities:

- Probability of having PKU (pp) = 1/4
- Probability of having albinism (aa) = 1/4

The combined probability of having both PKU and albinism:

Probability of pp and aa = 1/4 × 1/4 = 1/16

Therefore, the chance of having a child with both PKU and albinism is 1/16 or 6.25%.

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**12.2** **For any gene with a dominant allele C and recessive allele c, what proportions of the offspring from a CC X Cc cross expected to homozygous dominant, homozygous recessive and heterozygous?**

**Sol.** In a cross between CC (homozygous dominant) and Cc (heterozygous), the offspring inherit one allele from each parent:

1. **Homozygous dominant (CC):** Offspring inheriting the dominant allele from both parents will constitute 50% of the progeny.
2. **Homozygous recessive (cc):** No offspring inheriting the recessive allele from the heterozygous and homozygous parent (cc) will constitute 0% of the progeny.
3. **Heterozygous (Cc):** Offspring inheriting one dominant and one recessive allele will constitute 50% of the progeny.

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**12.3** **Two tall, yellow seeded pea plants were crossed, and some dwarf, green seeded plants resulted. (a) What were the genotypes of the parent plants? (b) What possible genotypes might there be among the tall, yellow-seeded offspring?**

**Sol:** To solve this problem, we need to understand the inheritance of the traits in pea plants: height and seed color. We will assume tall (T) is dominant over dwarf (t), and yellow seeds (Y) are dominant over green seeds (y).

**(a): Genotypes of the Parent Plants**

The fact that crossing two tall, yellow-seeded pea plants resulted in some dwarf, green-seeded offspring indicates that both parents must be heterozygous for both traits. This is because the recessive traits (dwarf and green) can only appear if the offspring inherit recessive alleles from both parents.

Thus, the genotypes of the parent plants must be:

TtYy (heterozygous for both height and seed color)

**(b): Possible Genotypes Among the Tall, Yellow-Seeded Offspring**

To determine the possible genotypes among the tall, yellow-seeded offspring, we will list the combinations that result in the dominant phenotypes for both traits.

For the offspring to be tall, they must have at least one dominant allele (T). For the offspring to have yellow seeds, they must have at least one dominant allele (Y).

The following all possible combinations would result in tall (T) and yellow seeds (Y):

1. TTYy
2. TtYy
3. TtYY
4. TTYy
5. TTYy

These combinations are possible due to the independent assortment of the alleles during gamete formation and fertilization.

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**12.4 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 pant? (b) If it turns out to be tall and yellow seeded what is the chance that its genotype is TTYY.**

**Sol.** To solve this genetics problem, break it down step by step using the principles of probability and genetics.

**Part (a): Probability of a Tall, Green Seeded Plant**

Given a TtYy pea plant self-pollinates, we need to determine the probability of obtaining a tall, green-seeded plant from a random seed.

1. **Genotype Analysis**:
   - Tt: "T" (tall) is dominant over "t" (short).
   - Yy: "Y" (yellow seeds) is dominant over "y" (green seeds).
2. **Self-Pollination Outcomes**
   - When a TtYy plant self-pollinates, the possible genotypes for each gene pair can be determined using the laws of independent assortment.
3. **Genotype Probabilities**
   - For height (Tt):
     - Probability of TT = 1/4
     - Probability of Tt = 1/2
     - Probability of tt = 1/4
   - For seed color (Yy):
     - Probability of YY = 1/4
     - Probability of Yy = 1/2
     - Probability of yy = 1/4



4. **Phenotype Probabilities**
   - **Tall (T):** Genotypes TT or Tt = Probability of 3/4
   - **Green seeds (y):** Genotype yy = Probability of 1/4
5. **Combined Probability**:

To get a tall, green-seeded plant (Tall and yy):

$$P(\text{Tall, green}) = P(\text{Tall}) \times P(\text{green})$$

$$P(\text{Tall, green}) = 3/4 \times 1/4 = 3/16$$

So, the probability that the seed will produce a tall, green-seeded plant is 3/16.

**Part (b): Probability of Genotype TTYY Given Tall and Yellow-Seeded**

Given that the plant is tall and yellow-seeded, we need to determine the probability that its genotype is TTYY.

1. **Genotype Analysis for Tall and Yellow-Seeded:**
   - Tall: TT or Tt
   - Yellow seeds: YY or Yy
2. **Possible Genotypes for Tall and Yellow-Seeded Plants:**
   - TTYY
   - TTYy
   - TtYY
   - TtYy
3. **Probability of Each Genotype:**
   - P(TTYY) = 1/16
   - P(TTYy) = 2/16 = 1/8
   - P(TtYY) = 2/16 = 1/8
   - P(TtYy) = 4/16 = 1/4
4. **Combined Probability for Tall and Yellow-Seeded:**

$$P(\text{Tall, yellow}) = P(TTYY) + P(TTYy) + P(TtYY) + P(TtYy)$$

$$P(\text{Tall, yellow}) = \frac{1}{16} + \frac{2}{16} + \frac{2}{16} + \frac{4}{16} = \frac{9}{16}$$

5. **Conditional Probability:**
   To find the probability that the genotype is TTYY given that the plant is tall and yellow-seeded, we use conditional probability:

$$P(TTYY|\text{ Tall, yellow}) = \frac{P(TTYY)}{P(\text{Tall, yellow})}$$

$$P(TTYY|\text{ Tall, yellow}) = \frac{\frac{1}{16}}{\frac{9}{16}} = \frac{1}{9}$$

So, the probability that a tall and yellnw-seeded plant has the genotype TTYY is $\frac{1}{9}$.

**Hypothesize that in a dihybrid inheritance pattern of colour and texture of pea seed, the two traits are not interdependent.**

**Observation and Background**
- It has been observed that the color and texture of pea seeds are inherited independently of each other.

**Hypothesis Statement**
- We hypothesize that in a dihybrid inheritance pattern, the traits of pea seed color and texture are not interdependent.

**Explanation Based on Chromosomal Behavior**
- The independence of these traits can be explained by the behavior of chromosomes during meiosis.
- Independent assortment, a key mechanism in meiosis, implies that the genes for these traits are located on different chromosomes.

**Genetic Location of Traits**
- Specifically, the alleles responsible for seed color are found on one chromosome pair, while the alleles for seed texture are located on a different chromosome pair.

**Conclusion**
- This arrangement allows each trait to be inherited independently, leading to the observed patterns of diversity in the offspring.

## Test Your Skills

1. In Labrador Retrievers, coat color is determined by two genes. The first gene (B) determines black (B) or brown (b) coat color. The second gene (E) determines whether the coat color is expressed (E) or if the coat will be yellow (ee) regardless of the B gene. Eye color is determined by another gene, where brown eyes (E) are dominant to blue eyes (e). A black Labrador with genotype BbEe is crossed with a yellow Labrador with genotype bbee. What are the possible coat colors and eye colors of the puppies?
2. In addition to seed shape and color, Mendel studied the inheritance of flower position. Axial flowers (A) are dominant to terminal flowers (a). A plant heterozygous for seed shape (Rr), seed color (Yy), and flower position (Aa) is crossed with another plant that is heterozygous for all three traits (RrYyAa). What are the possible phenotypes and their expected proportions in the offspring?
3. In humans, the ABO blood group is determined by one gene with three alleles (IA, IB, i), where IA and IB are co-dominant, and i is recessive. Another gene determines eye color, where brown eyes (B) are dominant over blue eyes (b). A woman with blood type AB and heterozygous for brown eyes (IAIBBb) marries a man with blood type O and blue eyes (ii bb). What are the possible blood types and eye colors of their children?
4. In pea plants, round seed shape (R) is dominant to wrinkled seed shape (r), and yellow seed color (Y) is dominant to green seed color (y). A plant that is heterozygous for both traits (RrYy) is crossed with another plant that is also heterozygous for both traits (RrYy). What are the possible genotypes and phenotypes of the offspring? What is the expected phenotypic ratio?

## Multiple Choice Questions

### Law of Independent Assortment

**1. The law of independent assortment states that:**
A. Alleles of different genes separate independently during gamete formation.
B. Each pair of alleles assort independently during gamete formation.
C. Alleles of the same gene separate during gamete formation.
D. Each allele pair replicates independently during cell division.

**2. During which phase of meiosis does independent assortment occur?**
A. Prophase I
B. Anaphase II
C. Metaphase I
D. Telophase I

**3. How many possible combinations of chromosomes can human gametes have due to independent assortment?**
A. 23
B. 46
C. 8 million
D. 23 million

**4. What does genetic linkage affect?**
A. Independent assortment of genes
B. Mutation rate of genes
C. Assortment of genes located on the same chromosome
D. The number of chromosomes in a cell

**5. How are sex-linked traits inherited differently in males?**
A. Males inherit more genes on the X chromosome.
B. Males are more prone to express recessive X-linked traits.
C. Males inherit X-linked traits from their fathers.
D. Males can inherit two copies of the X chromosome.

**6. What role does independent assortment play in evolution?**
A. Decreases genetic diversity
B. Contributes to genetic variation and adaptation
C. Ensures genes are inherited together
D. Reduces the effectiveness of natural selection

**7. What is the significance of crossing over in genetic variation?**
A. It decreases the number of chromosomes.
B. It creates new combinations of genes on the same chromosome.
C. It ensures identical inheritance of genes.
D. It reduces the number of possible gametes.

**8. Which Mendelian principle explains the behavior of chromosomes during meiosis?**
A. Law of Dominance
B. Law of Independent Assortment
C. Law of Uniformity
D. Law of Segregation

**9. Mathematically, the probability (P) is equal to:**
A. $P = a \times n$
B. $P = a + n$
C. $P = a/n$
D. $P = a - n$

**10. What is the probability of offspring inheriting a recessive phenotype in the F2 generation of a monohybrid cross?**
A. 0%
B. 25%
C. 50%
D. 75%

## Answer Key

| 1. B | 2. C | 3. C | 4. C | 5. B | 6. B | 7. B | 8. B | 9. C | 10. D |
|---|---|---|---|---|---|---|---|---|---|

**SLO 12.7 to 12.10** — Exceptions to Mendelian Inheritance

Students will be able to:

12.7 Describe the exceptions to the Mendel's laws of inheritance.
12.8 Explain incomplete dominance and exemplify it through the inheritance of flower color in the 4 O'clock plant.
12.9 Differentiate between incomplete dominance and co-dominance.
12.10 Define alleles and multiple alleles.



# EXCEPTIONS TO MENDELIAN INHERITANCE

- Mendel's laws of inheritance describe how traits are passed from parents to offspring through **dominant and recessive** alleles.
- However, there are several exceptions to these rules known as non-Mendelian inheritance patterns.
- These exceptions occur due to different ways in which alleles interact at the same genetic locus, especially in heterozygous conditions. Some key examples are:

1. **Incomplete Dominance**
   Unlike Mendel's observation where one allele completely dominates over another, incomplete dominance occurs when neither allele can fully dominate the other. As a result, the phenotype of the offspring is a blend or mixture of both parental traits.
   For example, when a red-flowered plant is crossed with a white-flowered plant, the result may be pink flowers.
2. **Codominance**
   In codominance, both alleles at a locus are expressed equally, and the phenotype displays characteristics of both alleles simultaneously.
   A classic example is the **ABO blood group** system, where both A and B alleles are fully expressed in individuals with AB blood type.
3. **Multiple Alleles**
   While Mendel worked with genes that existed in only two allelic forms, many genes exist in more than two forms, which is known as multiple alleles.
   The human ABO blood group system is also an example of this. In this blood group, three alleles (A, B, O) influence the blood type.

## Incomplete Dominance

- Incomplete dominance is a pattern of genetic inheritance where neither allele in a pair fully dominates the other.
- It results in an offspring with a phenotype that is intermediate between the two parents.
- The phenomenon of incomplete dominance was first documented by the German botanist **Carl Correns** in **1899**.

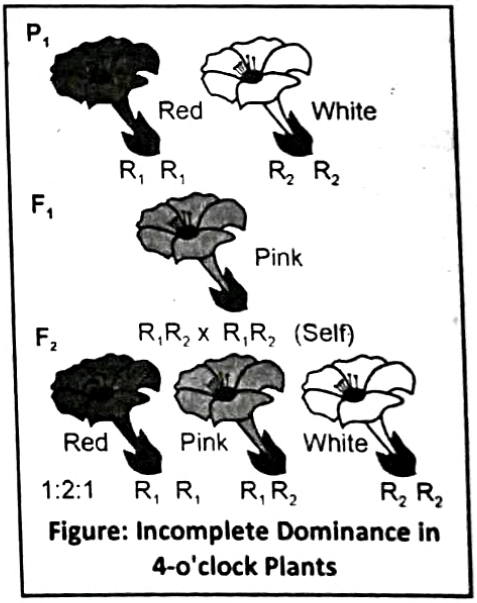


**Figure: Incomplete Dominance in 4-o'clock Plants**

### Key Concepts in Incomplete Dominance

- **Definition**: Incomplete dominance occurs when the phenotypic expression of the offspring's genotype is a blend of the parents' phenotypes.
- **Example**: A classic example is seen in the flower color of the Japanese four o'clock plant (*Mirabilis jalapa*). When a red-flowered plant is crossed with a white-flowered plant, the offspring have pink flowers. This shows that neither the red nor white allele is completely dominant.
- **Genetic Ratios in Offspring**: When pink-flowered plants from the F1 generation are cross-pollinated, their offspring exhibit red, pink, and white flowers in a 1:2:1 ratio. This ratio reflects that pink-flowered plants are heterozygous, possessing one red allele and one white allele, with neither allele being completely dominant over the other.

**Genetic Problem 12.5**

When a pink flower of the four O'clock plant is crossed with a red flower plant, what is the probability of:

(a) a red flower plant

(b) a pink flower plant

(c) the ratio of pink flower to red flower plant?

**Solution:**

The four o'clock plant (Mirabilis jalapa) exhibits incomplete dominance, where neither allele is completely dominant over the other. In this case, the red flower allele (R) and the white flower allele (W) result in a pink flower when they are heterozygous (RW).



**(a)** Probability of a red flower plant (RR):

Offspring will inherit one allele from each parent.

The red flower parent (RR) can only pass on the R allele.

The pink flower parent (Rr) can pass on either the R or r allele.

Therefore, the probability of a red flower (RR) offspring is 50%.

**(b)** Probability of a pink flower plant (Rr):

The pink flower parent (Rr) can pass on either the R or r allele.

The red flower parent (RR) only passes on the R allele.

Therefore, the probability of a pink flower (Rr) offspring is 50%.

**(c)** Ratio of pink flower to red flower plants:

The ratio of pink flower (Rr) to red flower (RR) plants in the offspring is 1:1.

This is because each parent contributes one allele, resulting in equal chances for each genotype combination (RR and Rr).

## Co-dominance

- Co-dominance is a type of genetic inheritance where both alleles at a gene locus are expressed equally and independently, resulting in a phenotype that displays characteristics of both alleles.
- This inheritance maintains and visibly expresses genetic diversity in populations.

**Science Titbits**

When the dominance is not complete, the capital case and small case letters will not be used to represent the genes, instead, only capital case letter differentiated by numeric figures will be used to represent the phenotypes.

### Characteristics of Co-dominance

- **Definition**: In co-dominance, both alleles at the same genetic locus are expressed without influencing each other. This results in a phenotype where traits from both alleles are visibly manifested.
- **Example of Co-dominance**: The human MN blood group system illustrates co-dominance clearly. In this system, the alleles $L^M$ and $L^N$ code for two different molecules on the surface of red blood cells, namely M and N molecules.
  - Individuals homozygous for the $L^M$ allele ($L^ML^M$) have red blood cells displaying only M molecules.
  - Individuals homozygous for the $L^N$ allele ($L^NL^N$) exhibit red blood cells with only N molecules.
  - Individuals heterozygous for the M and N alleles ($L^ML^N$) have red blood cells that carry both M and N molecules, clearly showing the independent expression of both alleles.

**Table - MN blood groups system in example of co-dominance**

| Blood Group phenotype | Antigen | Genotype |
|---|---|---|
| M blood | M antigen | $L^ML^M$ |
| N blood | N antigen | $L^NL^N$ |
| MN blood | Both antigen | $L^ML^N$ |

**Evaluate incomplete and co-dominance as variations of Mendel's research.**

Many patterns of inheritance which cannot be explained on the basis of Mendel's laws alone were discovered in plants and animals. Such patterns of inheritance are described as non-Mendelian inheritance. Incomplete dominance is a type of interaction where both the alleles of a given trait express as a blend (mixture) as against a normal Mendelian pattern where one allele is dominant over the other. As a result of this blending, an intermediate character is expressed. Co-dominance represents a situation where two allelic genes when present together in an individual, express their traits independently instead of showing a typical dominant recessive relationship. As a result the heterozygous progeny of the $F_2$ generation shows a phenotype that is different from both the homozygous parents.

The following table highlights the key differences in how alleles behave in heterozygous states under incomplete dominance and co-dominance.

**Table - Difference between Incomplete Dominance and Co-dominance**

| Characteristic | Incomplete Dominance | Co-Dominance |
|---|---|---|
| Allele Interaction in Heterozygotes | Both genes blend their phenotypic effects. | Both genes independently express their phenotypic effects. |
| Phenotype of Heterozygotes | Shows an intermediate phenotype between the two parental phenotypes. | Shows both parental phenotypes simultaneously. |
| Examples | Flower color of the 4 O'clock plant. | Human MN blood group and AB blood groups. |

**Evaluate Incomplete and Co-dominance as Variations of Mendel's Research**

Non-Mendelian inheritance refers to genetic patterns that do not conform to Mendel's laws of inheritance. These patterns, observed in plants and animals, involve more complex interactions between alleles than simple dominance or recessiveness.

**Incomplete Dominance**

- **Nature of Interaction**: Incomplete dominance occurs when neither allele in a pair is completely dominant over the other.
- **Phenotypic Outcome**: The result of this allele interaction is a phenotype that blends the traits of both alleles. For example, if a red-flowered plant is crossed with a white-flowered one, the offspring may have pink flowers, exhibiting an intermediate trait.

**Co-Dominance**

- **Nature of Interaction**: Co-dominance happens when both alleles at a locus are expressed equally in a heterozygous individual.
- **Phenotypic Outcome**: Unlike incomplete dominance, co-dominance allows both traits to appear simultaneously and independently. An example is seen in the human ABO blood group system, where both A and B alleles are fully expressed in individuals with AB blood type.

These variations from Mendel's research show that genetic inheritance can involve interactions that produce phenotypes blending both parents' traits or expressing both traits distinctly, contributing to the diversity seen in biological organisms.

## Gene and Allele

- **Gene** A gene is a segment of DNA that is located on a chromosome and encodes specific genetic information for particular traits.
- **Allele** Alleles are different forms of a gene that are found at the same location (locus) on homologous chromosomes. They can influence variations in the expression of a genetic trait.

### Multiple Alleles

**Definition**

- When a gene exists in more than two allelic forms, it is said to have **multiple alleles.**

**Production**

- Multiple alleles often result from gene mutations over time.

**Location**

- Despite the variety, all alleles of a gene occupy the same locus on a chromosome.

**Expression in Individuals**

- Although a gene may have many alleles, any diploid organism (like humans) can carry only two alleles at each locus — one from each parent. Haploid cells, such as gametes (sperm and egg cells), contain only one allele per locus.

**Example of Multiple Alleles**

- The ABO blood group system in humans exemplifies multiple alleles with three main forms—A, B, and O. This means a person can have combinations such as AA, AO, BB, BO, AB, or OO, but only two alleles can be present in any individual.

**Test Your Skills**

1. In the MN blood group system, the alleles $L^M$ and $L^N$ exhibit co-dominance. This means that individuals can be of type M (having $L^M L^M$), type N (having $L^N L^N$), or type MN (having $L^M L^N$).A man with type M blood ($L^M L^M$) marries a woman with type MN blood ($L^M L^N$).What are the possible blood types of their children? What is the expected ratio of these blood types?
2. A farmer has a herd of cattle with two coat color phenotypes: red and roan (a mix of red and white). If two roan cattle are crossed, what are the expected coat colors of their offspring?
3. Why might codominant alleles both be fully expressed in a heterozygous organism?
4. How does the ABO blood group system in humans demonstrate codominance?

## Multiple Choice Questions

### Exceptions to Mendelian Inheritance

**1. In incomplete dominance:**
A. One allele masks the effect of another.
B. Neither allele fully dominates, resulting in a blend of both parental traits.
C. Both alleles are fully expressed.
D. One allele changes the function of another.

**2. Flower colour of four o'clock plant shows:**
A. Co-dominance
B. Incomplete dominance
C. Dominance
D. None of these

**3. Haploid cell contains:**
A. Only one allele
B. Only two alleles
C. Three alleles
D. Four alleles

**4. How many alleles influence the ABO blood group system?**
A. Two alleles
B. Three alleles
C. Four alleles
D. One allele

**5. Wha: genetic concept is demonstrated when a red flower is crossed with a white flower to produce pink flowers?**
A. Codominance
B. Multiple alleles
C. Incomplete dominance
D. Mendelian inheritance

**6. When pink-flowered plants are crossed, what is the phenotypic ratio of their offspring for flower color?**
A. 3:1
B. 1:2:1
C. 2:1:1
D. 1:1:2

**7. In the human MN blood group system, what does a heterozygous individual exhibit?**
A. Only M molecules
B. Only N molecules
C. Both M and N molecules
D. Neither M nor N molecules

**8. Which pattern of inheritance involves genes that are located on the same chromosome and typically do not assort independently?**
A. Incomplete dominance
B. Codominance
C. Genetic linkage
D. Multiple alleles

**9. What are multiple alleles?**
A. Alleles that cause mutations
B. Different forms of a gene at the same locus
C. Genes that express multiple traits
D. None of the above

**10. What outcome is expected when a man with type M blood ($L^M L^M$) marries a woman with type MN blood ($L^M L^N$)?**
A. All children will have type M blood.
B. All children will have type N blood.
C. Children will have type M or MN blood in equal ratios.
D. Children will have type N or MN blood in equal ratios.

**Answer Key**

| 1. B | 2. B | 3. A | 4. B | 5. C | 6. B | 7. C | 8. C | 9. B | 10. C |
|---|---|---|---|---|---|---|---|---|---|

**SLO 12.11 to 12.19** — Blood Group Systems

Students will be able to:

12.11 State the alleles responsible for the trait of ABO blood groups.
12.12 Explain the case where two alleles have equal dominance through the genetics of the human blood group AB.
12.13 Name the various human blood group systems.
12.14 Investigate the reasons of $O^-$ (O negative) individual as the universal donor and $AB^+$ (AB positive) as the universal recipient.
12.15 Describe the occurrence of some other blood group systems.
12.16 Associate the positive and negative blood groups with the presence and absence of Rh factor.
12.17 Justify why Rh incompatibility could be a danger to the developing fetus and mother.
12.18 Explain Erythroblastosis foetalis in the light of antigen-antibody reaction.
12.19 Suggest measures to counter the problem of Erythroblastosis before it occurs.

# BLOOD GROUP SYSTEMS

- Blood group systems are classifications based on the presence or absence of specific molecules called **antigens** on the surface of red blood cells.
- These antigens, which are usually **glycoprotein molecules**, determine an individual's blood type.

## Major Blood Group Systems

- The International Society of Blood Transfusion recognizes up to 30 major blood group systems.

### ABO and Rh Systems

- The ABO and Rh (Rhesus) blood group systems are the most significant blood groups.
- These blood groups are important because if donor and recipient blood types are not compatible during blood transfusions, life-threatening complications can occur.

## Minor Blood Group Systems

- In addition to the ABO and Rh systems, there are over **two hundred** minor blood group systems.

### Significance of Minor Groups

- These minor systems generally do not cause complications in blood transfusions and are considered rare blood types.
- An example of a minor system is the MN blood group system, which is an example of co-dominance.

## ABO Blood Group System

- The ABO blood group system was discovered by Karl Landsteiner in 1901
- It is a classic example of multiple alleles in human genetics.
- This system is also present in various other primates, such as apes, chimpanzees, bonobos, and gorillas.
- The discovery of the ABO system marked a significant advancement in our understanding of human blood types and their genetic basis.

## Antigens of the ABO Blood Group System

- The ABO blood group system is characterized by the presence of specific antigens on the surface of red blood cells (RBCs).
- These antigens are glycoproteins and their presence or absence determines blood types:
  - **A Antigen**: When the A antigen is present on the RBCs, the blood group is classified as Type A.
  - **B Antigen**: The presence of the B antigen on RBCs defines the blood group as Type B.
  - **Both A and B Antigens**: If both A and B antigens are present, the blood group is Type AB, demonstrating co-dominance.

- **No A and B Antigens**: If neither the A nor B antigens are present, the blood group is Type O, indicating the absence of these specific antigens.

- This system of antigens is essential for determining compatibility in blood transfusions and has implications in medical treatments and genetic studies.



## Genetic Basis of the ABO Blood Group System

- The ABO blood group is determined by a single autosomal gene known as the isohaemagglutinogen gene.
- It is represented by the symbol 'I'.
- This gene has three different alleles that control the blood group:
  - **$I^A$ Allele**: Responsible for producing the A antigen on red blood cells.
  - **$I^B$ Allele**: Responsible for producing the B antigen on red blood cells.
  - **i Allele**: Does not produce either A or B antigens, resulting in the O blood type.

**Science Titbits**

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" present on chromosome 19.

### Dominance Relationships

- **Dominance over i**: Both $I^A$ and $I^B$ alleles are completely dominant over the i allele. This means that the presence of either $I^A$ or $I^B$ will mask the effect of i.
- **Co-dominance of $I^A$ and $I^B$**: The $I^A$ and $I^B$ alleles are co-dominant to each other. When an individual inherits one of each of these alleles (e.g., $I^A I^B$), both antigens are equally expressed, resulting in the AB blood type.

### Expression throughout Life

- **Continuous Expression**: The expression of these blood group alleles begins at an early embryonic stage and continues throughout a person's life.
- **Consistency of Phenotype**: Due to the constant expression of these alleles, the blood group phenotype of an individual remains unchanged from birth until death.

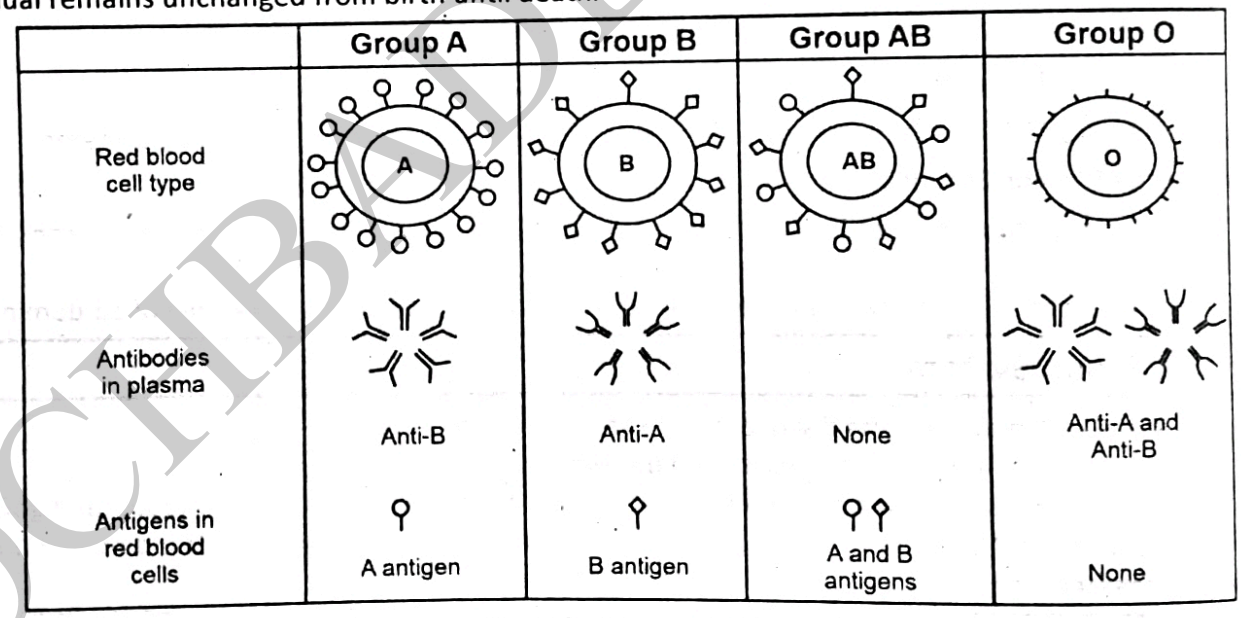



**Figure: Multiple Alleles and ABO Blood Groups**

**Table: Multiple Alleles and ABO Blood Groups**

| Blood Group (phenotype) | Antigen | Genotypes | Antibodies | Transfusions |
|---|---|---|---|---|
| A | A | $I^A I^A$ or $I^A i$ | B | A and O |
| B | B | $I^B I^B$ or $I^B i$ | A | B and O |
| AB | Both | $I^A I^B$ | None | Any |
| O | None | $ii$ | Both | Only O |

## Antibodies in the ABO Blood Group System

- In the ABO blood group system, the presence of specific antibodies plays a crucial role in determining the safety and compatibility of blood transfusions.
- These antibodies are naturally occurring and are produced against the ABO antigens that an individual does not possess.

### Function and Reaction

- If a person receives blood containing antigens that do not match their own, their immune system will react.
- The antibodies in the recipient's blood will bind to these foreign antigens, causing **agglutination (clumping)** and potentially causing dangerous reactions.



### Antibody Production

- **Type A Blood**: Individuals have antibodies against B antigen (anti-B) in their plasma.
- **Type B Blood**: Individuals produce antibodies against A antigen (anti-A).
- **Type AB Blood**: People with AB blood do not produce anti-A or anti-B antibodies, as they naturally have both A and B antigens.
- **Type O Blood**: Individuals have neither A nor B antigens but have both anti-A and anti-B antibodies.

### Onset and Duration of Antibody Production

- **Early Development**: The production of these antibodies starts in embryonic life.
- **Lifelong Presence**: These antibodies continue to be produced throughout an individual's life without the need for any external stimulus.

## Overview of Human Blood Group Systems

- The International Society of Blood Transfusion recognizes 33 blood group systems comprising over 300 antigens.
- Some key blood group systems include ABO, MNS, Rhesus (Rh), Lutheran (LU), Kell (KELL), Lewis (LE), Duffy (FY), and Kidd (JK).
- Each of these systems plays a crucial role in medical contexts like transfusion and transplantation due to their unique antigens and antibodies.

**H-antigen**

H-antigen is the precursor to the ABO blood group antigens. It is present in all RBCs irrespective of the ABO system. Persons with the rare Bombay phenotype are homozygous for the H gene (HH), do not express H-antigen on their RBCs. As H antigen acts as precursor, its absence means the absence of antigen A and B. However, the individuals produce isoantibodies to H-antigen as well as to antigens A and B.

### ABO System

- The ABO system is the most important for transfusions and transplants as it includes significant antibodies (anti-A and/or anti-B) that are naturally present in individuals older than 6 months.

### Rhesus System

- The Rhesus system is the second most critical blood group after ABO.
- Anti-D immunoglobulin is administered to prevent Rh immunization in Rh-negative mothers after giving birth to an Rh-positive child.

### MNS System

- It is governed by an autosomal locus on chromosome 4 with co-dominant alleles $L^M$ and $L^N$.
- Anti-M and anti-N antibodies are typically IgM types, which rarely cause transfusion reactions.

### Lutheran System

- It consists of four pairs of allelic antigens. Each represents a single amino acid substitution on the Lutheran glycoprotein located on chromosome 19.
- Antibodies against this group are rare and not usually clinically significant.

### Kell System

- It is identified by an immune antibody, anti-K. It was first noticed in Mrs. Kellacher's serum reacting to her newborn's erythrocytes.
- There are now 25 known Kell antigens. Anti-K antibodies can cause severe hemolytic disease of the fetus and newborn (HDFN) and hemolytic transfusion reactions (HTR).

### Duffy System

- The Duffy antigen was first isolated in a patient named Duffy who had hemophilia.
- Fya and Fyb antigens on the Duffy glycoprotein result in four possible phenotypes.
- The antibodies, which are IgG subtypes, can cause hemolytic transfusion reactions.

### Kidd System

- The Kidd antigen (Jk) is a glycoprotein on RBC membranes that acts as a urea transporter in RBCs and renal endothelial cells.
- Kidd antibodies are rare but can cause severe transfusion reactions.



## Transfusion Principles

- Transfer of blood from a donor to a recipient is called **transfusions**. For a safe transfusion, it is important to match the donor's blood group antigens with the recipient's antibodies to prevent adverse reactions.

### Agglutination and Compatibility

- This occurs when the antigens of the donor blood react with the antibodies (**agglutinins**) of the recipient, causing the red blood cells to clump together. This reaction can lead to serious complications.

### Blood Group Compatibility

#### Type A Blood Group

- Individuals with type A blood can receive blood from type A or type O donors. They have anti-B antibodies, which means they cannot receive blood that carries the B antigen.

#### Type B Blood Group

- Those with type B blood can receive from type B or type O donors. They have anti-A antibodies and cannot accept blood with the A antigen.

#### Type AB Blood Group

- Individuals with type AB blood can receive blood from any ABO group (A, B, AB, or O) because they do not have any ABO antibodies. This makes them universal recipients.

#### Type O Blood Group

- People with type O blood can only receive from type O donors because they have both anti-A and anti-B antibodies, which react against blood containing either A or B antigens. However, because type O blood lacks A and B antigens, it can be donated to any ABO blood group, making type O individuals universal donors.

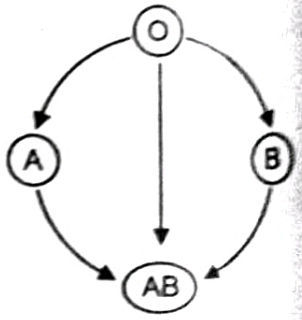


**Figure: Blood Transfusion Model**

### Special Considerations for Rh Factor

#### Rh-Negative

- Individuals with Rh-negative blood must receive Rh-negative blood only to avoid immunological reactions. However, they can donate to Rh-positive or Rh-negative recipients, making Rh-negative donors particularly valuable.

**Test Your Skills**

1. How do antigens on the surface of red blood cells determine blood types?
2. What are minor blood group systems, and how do they differ from major systems?
3. Give an example of a minor blood group system and explain its clinical significance.
4. Why individuals with Type O blood are considered universal donors?
5. A newborn has jaundice and is found to have anti-K antibodies in their blood. Which blood group system is involved, and what might be the cause?
6. A patient with Type B blood receives a transfusion from a Type A donor. Explain the immunological response that would occur.
7. How do the naturally occurring anti-A antibodies in the patient's blood contribute to this reaction?

**Genetic Problem 12.6**

**A person has type A blood group while his wife has type B, they have four children each with different blood group in the ABO system. Explain how it is possible.**

**Solution:** It is possible for a person with blood type A and a person with blood type B to have four children each with different blood groups (A, B, AB, O) due to the genetic variability in ABO blood group inheritance. Each parent contributes one allele (A or B) to their offspring, resulting in diverse combinations:

Child 1 inherits AA (blood type A) from parent A.
Child 2 inherits BB (blood type B) from parent B.
Child 3 inherits AB (blood type AB) through co-dominance.
Child 4 inherits OO (blood type O) from both parents' recessive alleles. This variation occurs because A and B alleles segregate independently during gamete formation and recombine randomly during fertilization.

**Genetic Problem 12.7**

**A woman with blood type B has a child with blood type 0. What are the genotypes of the mother and child? Which genotypes could the father not have?**

**Solution:**

1. Mother's Genotype (Blood type B): $I^bI^b$ or $I^bi$.
2. Child's Genotype (Blood type O): ii
3. Father's Genotypes: $I^ai$ (heterozygous) or ii ( for O).

- The father could not have the genotype $I^bI^b$ because in that case, all offspring would have blood type B ($I^ai$ or $I^bi$).



**Derive an idea to get alternatives of blood transfusion.**

Some approaches are available that can decrease the need for a blood transfusion. The options currently available are:

1. Volume expanders are used to prevent or treat the shock associated with loss of body fluids. The most common volume expanders used include salt water (normal saline) and saline with some chemicals added (Ringer's solution).
2. Hematopoietic growth factors encourage the bone marrow to make more red blood cells. These growth factors can be made in the laboratory and given to people with low blood cell counts.
3. Erythropoetin is a naturally occurring hormone produced by the kidneys. It stimulates the body to produce more red blood cells and is used to treat anaemia. It is widely used as a transfusion alternative.
4. Aprotinin is a drug that is given before heart surgery to reduce the risk of bleeding and the need for transfusion.

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

The cross between blood group O (ii) and O (ii) will have blood group O (ii) in all the offspring. The cross between heterozygous $A^A$ and $A^Ai$ will produce 25% offspring having blood group O (ii), and likewise heterozygous $\beta'$ and I $\beta_i$ will produce 25% offspring having blood group O (ii).Cross between heterozygous $A^A$ and $I^Bi$ will produce 25% offspring having blood group O (ii). That's why blood group allele ' ' is more frequent in the population. For example Australia 40%, Canada 39%, Iceland 47%, Ireland 47% UK 37%, USA 37%.

**Justify blood donation as a service to suffering humanity.**

Blood donation is a social responsibility. The donor is donating for it as it will be used in saving lives of human beings. As The Quran says in Surah 5 verse 32 "if anyone saves a life, it shall be as though he had saved the lives of all mankind." Millions of people owe their lives to people whom they will never know or meet in their lifetime. They are none other than those people, who have donated their blood freely and without any reward - voluntary blood donors. Voluntary unpaid donors are the foundation of a safe blood supply which saves millions of human beings from the jaws of untimely death. Blood donation is a noble, selfless service. It gives the donor a feeling of joy and contentment. Also this is an expression of love for Mankind, as blood knows no caste, colour, creed, religion or race, country, continent or sex. Do you know that 'one unit of blood can save three lives rather than

## Rh Blood Group System Overview

- The Rh blood group system is the second most clinically significant blood group system after ABO.
- It was named after its initial discovery in the **Rhesus monkey** by **Karl Landsteiner** in the 1930s.

### Antigens of the Rh Blood Group System

#### Presence of D Antigen

- This system is primarily defined by the presence or absence of the **D antigen**, also known as the **Rh factor**.
  - Rh Positive: Individuals who have the D antigen on their red blood cells (RBCs) are referred to as Rh positive.
  - Rh Negative: Those lacking the D antigen are Rh negative.

#### Clinical Implications

- The presence or absence of the D antigen is crucial in blood transfusions and is a significant factor in hemolytic disease of the newborn, also known as erythroblastosis fetalis.



### Genetic Basis of the Rh Blood Group System

- The presence of the D antigen is controlled by the D gene.

#### Dominance

- The D allele is completely dominant over its alternative allele d.

#### Genotypes and Phenotypes:

- Individuals with DD or Dd genotypes possess the D antigen and are Rh-positive.
- Individuals with the dd genotype do not have the D antigen and are Rh-negative.

### Implications for Blood Transfusion

#### Universal Donor

- The O negative ($O^{-ve}$) blood type is considered the actual universal donor because it lacks both ABO and Rh antigens, minimizing the risk of antigen-antibody reactions during transfusions.

#### Universal Recipient

- The AB-positive ($AB^{+ve}$) blood type is regarded as the actual universal recipient because it lacks anti-A, anti-B, and anti-Rh antibodies, allowing it to receive blood from any donor without resistance.

**Table: Rh-Blood groups system**

| Blood group (phenotype) | Rh Antigen/factor | Genotypes | Anti-Rh-Antibody | Transfusions |
|---|---|---|---|---|
| $Rh^{+ve}$ | Present | $DD$ or $Dd$ | Not produced | $Rh^{+ve}, Rh^{-ve}$ |
| $Rh^{-ve}$ | Absent | $dd$ | Produced (if stimulated) | $Rh^{-ve}$ |

## Anti-Rh Antibody and Transfusion Principles

- The Rh blood group system includes a specific response mechanism for the production of anti-Rh antibodies, which is significant in the context of blood transfusions, especially for Rh-negative individuals.

### Production of Anti-Rh Antibodies

#### Stimulus Requirement

- Unlike antibodies in the ABO system, which are naturally occurring, the production of anti-Rh antibodies in Rh-negative individuals requires a specific stimulus—exposure to Rh-positive blood.

#### Mechanism

- When an Rh-negative individual receives blood containing the Rh antigen (from an Rh-positive donor), their immune system recognizes these antigens as foreign and starts producing anti-Rh antibodies.

### Implications for Blood Transfusion

#### Compatibility Issues

- Rh-positive blood is completely incompatible with Rh-negative recipients. The exposure of Rh-negative individuals to Rh-positive blood triggers the production of anti-Rh antibodies, leading to potential immune reactions.

#### Long-term Response

- Once an Rh-negative individual is exposed to Rh antigens and begins producing anti-Rh antibodies, this immune response continues for their entire life. Subsequent exposures to Rh-positive blood can result in more severe immune reactions.

#### Safe Transfusions for Rh-Positive Recipients

- Rh-negative blood from donors who have never been exposed to Rh antigens (and thus do not have anti-Rh antibodies) can be safely transfused to Rh-positive recipients, as there is no risk of antibody-mediated reactions.

## Erythroblastosis Fetalis

- **Erythroblastosis fetalis** is a severe blood disorder that affects fetuses due to incompatibility between maternal and fetal blood.

### Problems and Complications in Fetuses Due to Rh Incompatibility

- Erythroblastosis fetalis can lead to severe complications in fetuses when maternal anti-Rh antibodies penetrate the placenta and enter the fetal bloodstream.

### Hemolysis and Its Effects

#### Initiation of Hemolysis

- The anti-Rh antibodies from the mother cause the breakdown of fetal red blood cells (hemolysis).

#### Development of Anemia

- As red blood cells are destroyed, the fetus becomes anemic.

#### Release of Erythroblasts

- The anemic fetus compensates by releasing a large number of immature red blood cells (erythroblasts) into the bloodstream, a condition named erythroblastosis fetalis.

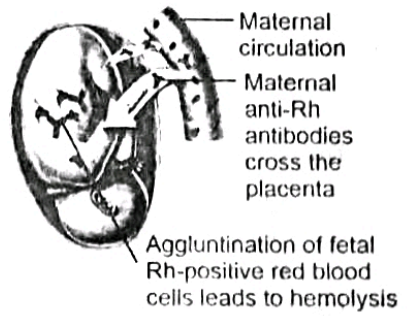



**Figure: Maternal-Foetal Rh-Incompatibility**



### Secondary Complications

#### Organ Enlargement

- Continued anemia leads to rapid production of red blood cells by the fetus's liver and spleen, causing these organs to enlarge.

#### Bilirubin Accumulation

- The breakdown of red blood cells produces bilirubin. High levels of bilirubin in the fetus can lead to brain damage and jaundice, evident by the yellowing of the skin and the whites of the eyes.

### Post-birth Risks and Interventions

#### Risk at Birth

- If the baby is born alive, it may suffer from severe hemolytic anemia and jaundice.

#### Immediate Medical Intervention

- It is critical to immediately replace the baby's blood with Rh-negative blood that does not contain anti-Rh antibodies to mitigate further health issues.

## Causes and Risk Factors of Erythroblastosis Fetalis

- Erythroblastosis fetalis primarily occurs due to Rh incompatibility between an Rh-negative mother and her Rh-positive fetus, a condition influenced by the genetic contributions of both parents.



### Genetic Contributions and Risk Assessment

#### Maternal-Fetal Rh Incompatibility

- The condition most commonly arises when an Rh-negative woman conceives a baby with Rh positive blood, typically when the father is Rh-positive.

#### Father's Genotype Influence

- **Homozygous Dominant (DD)**: If the father's genotype is DD (homozygous dominant for the Rh factor), all offspring will inherit one Rh-positive allele (D) and will be Rh-positive (Dd).
- **Heterozygous (Dd)**: If the father's genotype is Dd, there is a 50% chance each child will inherit the Rh-positive allele (D) and be Rh-positive.

### Likelihood of Erythroblastosis Fetalis

- There is a persistent risk of erythroblastosis fetalis in any pregnancy where an Rh-negative mother conceives an Rh-positive fetus due to the potential development of maternal anti-Rh antibodies.

## Prevention of Erythroblastosis Fetalis

- Erythroblastosis fetalis, a condition caused by Rh incompatibility, can be effectively prevented through timely medical interventions.

### Rh Immunoglobulin (RhIg) Therapy

#### Role of RhIg (RhoGAM)

- Rh immunoglobulin (RhIg), commonly known as RhoGAM, is administered to prevent the pregnant woman from developing antibodies against Rh-positive blood.

**Science Titbits**

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

#### Mechanism

- The injection contains anti-Rh antibodies that neutralize any of the baby's Rh-positive red blood cells that might enter the mother's bloodstream before they can stimulate her immune system to produce her own antibodies.

#### Limitation

- This treatment is not effective for women who have already been sensitized to Rh-positive blood.

### Administration Schedule

- To maximize effectiveness, RhoGAM should be administered at specific times:

#### During Pregnancy

- At 28 weeks of gestation prevent the mother from becoming sensitized in the later stages of pregnancy.

#### After Delivery

- Within 72 hours after delivery if the newborn is Rh-positive, to prevent sensitization from any fetal blood cells that may have entered the mother's circulation.

#### After Pregnancy Loss

- Within 72 hours of a miscarriage, abortion, or ectopic pregnancy to prevent sensitization following these events.

#### After Invasive Procedures

- Following procedures that could potentially introduce fetal cells into maternal circulation, such as amniocentesis or chorionic villus sampling (CVS).

#### After Vaginal Bleeding

- Any occurrence of vaginal bleeding during pregnancy may warrant a dose of RhIg to prevent sensitization.

- Properly timed doses of RhoGAM are crucial in preventing the development of Rh antibodies in Rh-negative women, thereby significantly reducing the risk of erythroblastosis fetalis in current and future pregnancies.

**Test Your Skills**

1. A child born with erythroblastosis fetalis requires ongoing medical monitoring. What long-term complications might arise, and how should they be managed medically?
2. A couple is planning their second pregnancy. The mother is Rh-negative, and the father is Rh-positive (genotype Dd). Discuss the likelihood of erythroblastosis fetalis in their next child. What preventive measures should the couple consider before conception and during pregnancy?
3. What are the possible genotypes and corresponding phenotypes for Rh-positive and Rh-negative individuals?

### Genetic Problem 12.8

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, and what are the chances that their child will be affected with erythroblastosis foetalis?

1. Mother's Genotype: Rh-negative (dd)
2. Father's Genotype: Rh-positive (DD or Dd)
3. Child's Genotype: Possibilities depend on the father's genotype:
   - If the father is DD (homozygous Rh-positive), all children will be Rh-positive (Dd).
   - If the father is Dd (heterozygous Rh-positive), there's a 50% chance of having Rh-positive (Dd) children and 50% chance of Rh-negative (dd) children.
4. Risk of Erythroblastosis Fetalis:
   If the fetus inherits the Rh-positive allele (D) from the father and the mother is Rh-negative (dd), there is a risk of the mother developing antibodies against the Rh antigen in subsequent pregnancies, potentially causing erythroblastosis fetalis in Rh-positive fetuses.
5. The chances of a child being affected by erythroblastosis fetalis depend on the father's Rh genotype. With an Rh-positive father, there's a 50% chance that each child will be Rh-positive and at risk for the condition if the mother develops antibodies.



## Multiple Choice Questions

### Blood Group Systems

1. **What determines an individual's blood type?**
   A. Blood cells count
   B. Antigens on the surface of red blood cells
   C. Plasma composition
   D. White blood cell type
2. **How many major blood group systems does the International Society of Blood Transfusion recognize?**
   A. 15 B. 20
   C. 30 D. 33
3. **What are the most significant blood groups for blood transfusions?**
   A. MN and Lewis B. ABO and Rh
   C. Kell and Duffy D. Lutheran and Kidd
4. **What blood group system involves antigens like A and B?**
   A. Rh B. ABO
   C. MN D. Duffy
5. **Who discovered the ABO blood group system?**
   A. Watson and Crick
   B. Karl Landsteiner
   C. Gregor Mendel
   D. Charles Darwin
6. **What happens if donor and recipient blood types are not compatible during transfusion?**
   A. Nothing significant
   B. Slight discomfort
   C. Life-threatening complications
   D. Minor allergic reactions
7. **What is an example of a minor blood group system?**
   A. ABO B. Rh
   C. MN D. Duffy
8. **Which allele is not responsible for producing any antigens in the ABO blood group system?**
   A. IA B. IB
   C. i D. AB
9. **What does the 'i' allele produce?**
   A. A antigen B. B antigen
   C. Both A and B antigens D. No antigens

10. What kind of dominance relationship exists between IA and IB alleles?
A. Complete dominance B. Co-dominance
C. Incomplete dominance D. No dominance

11. What blood group can receive from any ABO group?
A. A B. B
C. O D. AB

12. What type of antibodies are present in individuals with Type O blood?
A. Anti-A and Anti-B B. Anti-A and Anti-B
C. Anti-AB D. None

13. When do ABO blood group antibodies begin to develop?
A. At birth B. In embryonic life
C. After one year D. During adolescence

14. What blood group is considered the universal donor?
A. A B. B
C. AB D. O

15. What antigens does a person with AB blood type have?
A. A only B. B only
C. Both A and B D. Neither A nor B

16. Which blood group system includes co-dominant alleles $L^M$ and $L^N$?
A. ABO B. Rh
C. MNS D. Duffy

17. What antibodies do individuals with type AB blood produce?
A. Anti-A only B. Anti-B only
C. Both Anti-A and Anti-B D. None

18. What is the primary clinical importance of the Rh blood group system?
A. Genetic diversity
B. Blood transfusion compatibility
C. Evolutionary biology
D. Cellular function

19. What genotype characterizes Rh-negative individuals?
A. DD B. Dd
C. dd D. Either DD or Dd

20. What happens when an Rh-negative individual receives Rh-positive blood?
A. No reaction
B. Production of anti-Rh antibodies
C. Increased blood pressure
D. Decreased immune response

21. How is Rh incompatibility primarily prevented?
A. Regular blood transfusions
B. Administration of Anti-D immunoglobulin
C. Genetic modification
D. Avoiding transfusions altogether

22. What type of antigen is involved in the Duffy blood group system?
A. ABO antigen B. Rh antigen
C. Fya and Fyb antigens D. M and N antigens

23. What blood type can receive blood from any donor in terms of ABO compatibility?
A. A B. B
C. O D. AB

24. What is the role of antibodies in the ABO blood group system?
A. Transport oxygen
B. Prevent incompatible blood transfusions
C. Carry nutrients
D. Regulate blood pressure

25. What is the significance of the anti-K antibody in the Kell blood group system?
A. It is insignificant
B. It is rarely produced
C. It can cause hemolytic disease of the fetus and newborn
D. It aids in oxygen transport

26. How are blood group systems relevant in medical contexts?
A. Only for academic purposes
B. For transfusions and transplants
C. For diagnosing all diseases
D. For cosmetic treatments

**Answer Key**

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1. B | 2. C | 3. B | 4. B | 5. B | 6. C | 7. C | 8. C | 9. D | 10. B | 11. D | 12. B | 13. B | 14. D | 15. C |
| 16. C | 17. D | 18. B | 19. C | 20. B | 21. B | 22. C | 23. D | 24. B | 25. C | 26. B | | | | |

**SLO 12.20 to 12.23 — Polygenic Inheritance and Epistasis**

Students will be able to:

12.20 Define and relate the terms polygenic and epistasis.

12.21 Describe polygenic inheritance using suitable examples from plants (grain colour in wheat) and animals (skin colour in man).

12.22 List at least five polygenic traits discovered in humans.

12.23 Give an example of epistasis from mammals (coat colour inheritance in Labrador retrievers) and one from plants (pigment phenotype in sweet pea, Lthyrus odoratus) and justify modified Mendelian ratios.

# POLYGENIC INHERITANCE AND EPISTASIS

- Polygenic inheritance describes how certain traits are controlled by multiple genes, often resulting in a continuous variation in phenotypes, such as height, weight, intelligence, and skin color in humans, or grain color in wheat.
- Some traits have a large number of alternative phenotypes that have small and less noticeable differences so they have continuous variations such as height, weight, intelligence and skin colour in human; and grain colour in wheat.
- Such traits 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 phenotypes.
- Such traits are encoded by alleles of two or more different gene pairs found at different loci, all influencing the same trait in an additive way i.e., the intensity of phenotype depends upon the number of particular effects causing alleles. These quantitative traits are therefore called polygenic traits.



### Characteristics of Polygenic Traits

#### Multiple Genes Involved

- Traits with a broad range of phenotypes, such as skin color or height, are typically influenced by several genes located at different genetic loci.

#### Additive Effects

- The phenotypic intensity of these traits depends on the cumulative effect of alleles from multiple genes. Each contributing gene adds a small effect that may enhance or diminish the trait.

#### Continuous Variation

- Unlike traits controlled by a single gene, polygenic traits do not exhibit distinct categories but instead show a range of outcomes. They form a continuous spectrum.

### Role of Polygenes

#### Definition of Polygenes

- The multiple genes that collectively influence a polygenic trait are called **polygenes**. Each polygene contributes a small, additive effect on the overall phenotype.

#### Summation of Effects

- The overall phenotype is the result of the summation of both positive and negative effects from all individual polygenes. This interaction creates the continuous variation in traits like height or skin color.

## Wheat Grain Color and Polygenic Inheritance

- Wheat grain color is as an illustrative example of polygenic inheritance, where multiple genes contribute to a trait resulting in a spectrum of observable phenotypes.

### Overview of Wheat Grain Color Variation

#### Continuous Variation

- Wheat grains display a range of color variations from white to dark red. It shows a continuous phenotypic variation.

#### Phenotypes

- Across global wheat populations, approximately seven distinct color phenotypes are identified, indicating a broad genetic diversity.

### Genetic Study and Findings

#### Initial Cross Study

- The genetics of wheat grain color was explored through a cross between a **homozygous dark red grain plant** and a **homozygous white grain plant**, resulting in all F1 generation grains displaying a light red color. This initially suggested incomplete dominance.

### F2 Generation Outcome

- Further crossing of F1 plants revealed the F2 generation exhibited exactly seven shades of color. This pattern confirmed the involvement of multiple genes.

## Genetic Mechanism Behind Color Variation

### Gene Pairs Involved

- Three gene pairs identified as Aa, Bb, and Cc at three different loci are responsible for the color of wheat grain.

### Allele Impact on Pigmentation

- **Positive Effect Alleles**: Alleles A, B, and C each contribute equally to the production of red pigment.
- **Negative Effect Alleles**: Alleles a, b, and c do not produce red pigment, diminishing the color intensity.

### Color Intensity Based on Allele Combination

- **Six Alleles for Red Pigment (AABBC):** Produces dark red grains.
- **No Alleles for Red Pigment (aabbcc):** Results in white grains.
- **Intermediate Combinations**: Various combinations of these alleles result in incremental shades ranging from light pink to moderately dark red.

## Factors Affecting Pigmentation

- Environmental conditions such as light exposure, water availability, and nutrient levels also significantly influence grain color, interacting with genetic factors to determine the final phenotype.

**Range of Phenotypes in Wheat Grain Colours**

| Pigment allele | 6 | 5 | 4 | 3 | 2 | 1 | 0 |
|---|---|---|---|---|---|---|---|
| Range of phenotypes | Dark red | Moderately dark red | Red | Light red | Pink | Light Pink | White |
| Ratio | 1 | 6 | 15 | 20 | 15 | 6 | 1 |

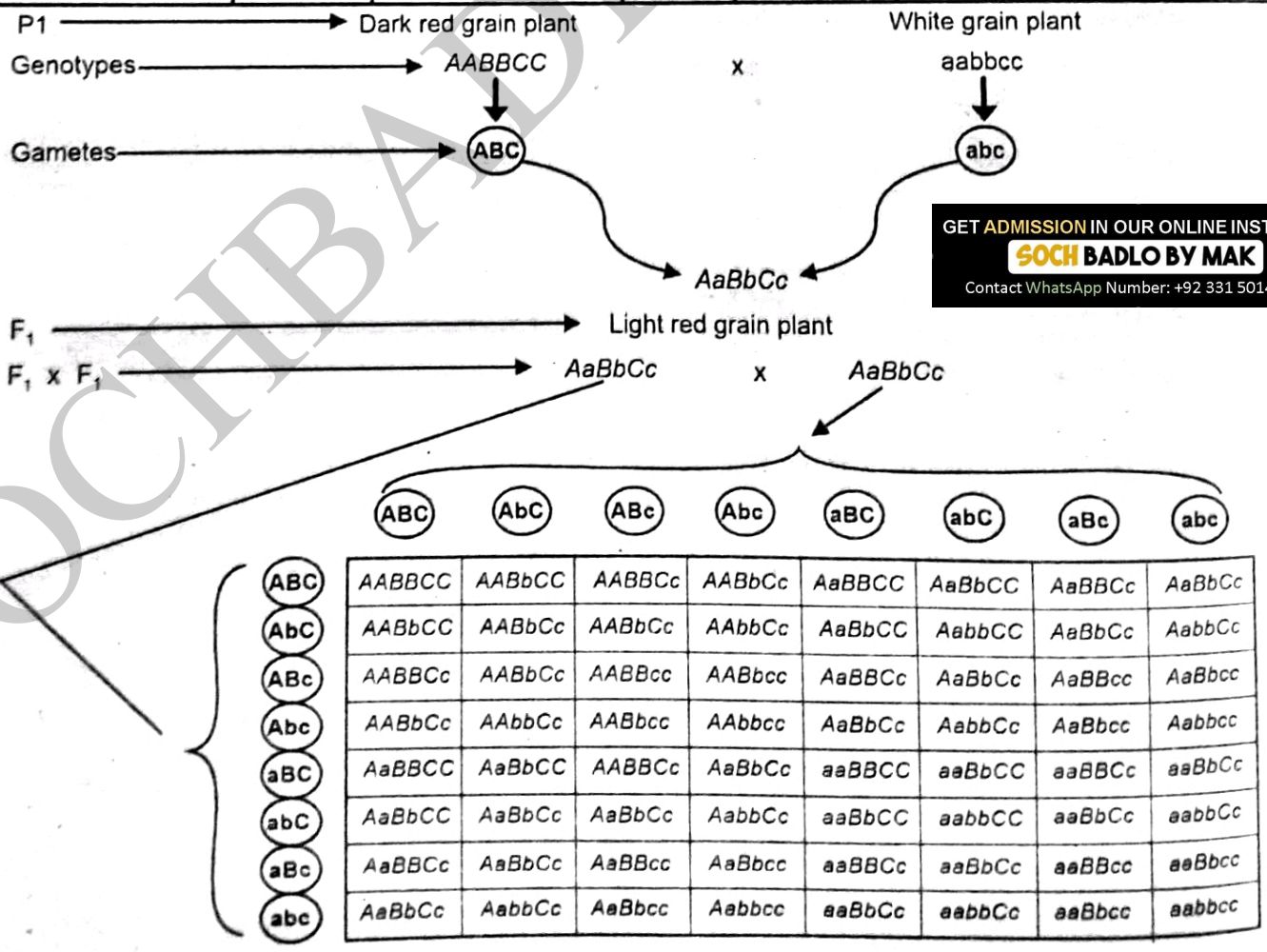



Figure: Inheritance of Wheat Grain Colour

# Inheritance of Human Skin Color

- Human skin color shows polygenic inheritance. Here multiple genes contribute to the phenotype. They influence the amount of melanin produced in the skin.

## Genetic Determinants of Skin Color

### Melanin Production

- Skin color variation primarily depends on the amount of melanin produced. Individuals with darker skin produce more melanin than those with lighter skin.

### Gene Regulation

- At least three genes, labeled as A, B, and C, are involved in regulating melanin production.

## Allelic Contributions to Skin Color

### Alleles for Dark Skin

- The alleles for darker skin are represented by uppercase letters (A, B, C). These alleles promote increased melanin production, contributing to darker skin tones.

### Alleles for Light Skin

- The alleles associated with lighter skin are denoted by lowercase letters (a, b, c). These alleles reduce melanin production, leading to lighter skin tones.

### Dominance and Phenotypic Expression

- There is no complete dominance among these alleles; heterozygous combinations (such as Aa, Bb, Cc) result in intermediate skin tones due to incomplete dominance.

## Phenotypic Range and Examples

### Skin Color Spectrum

- The interaction of these alleles results in at least seven different shades of skin color, ranging from very light (aabbcc) to very dark (AABBCC).

### Intermediate Phenotypes

- Most individuals possess an intermediate shade, such as the AaBbCc genotype, often observed in multiracial individuals like those of mixed African and European ancestry, sometimes referred to historically as mulatto.



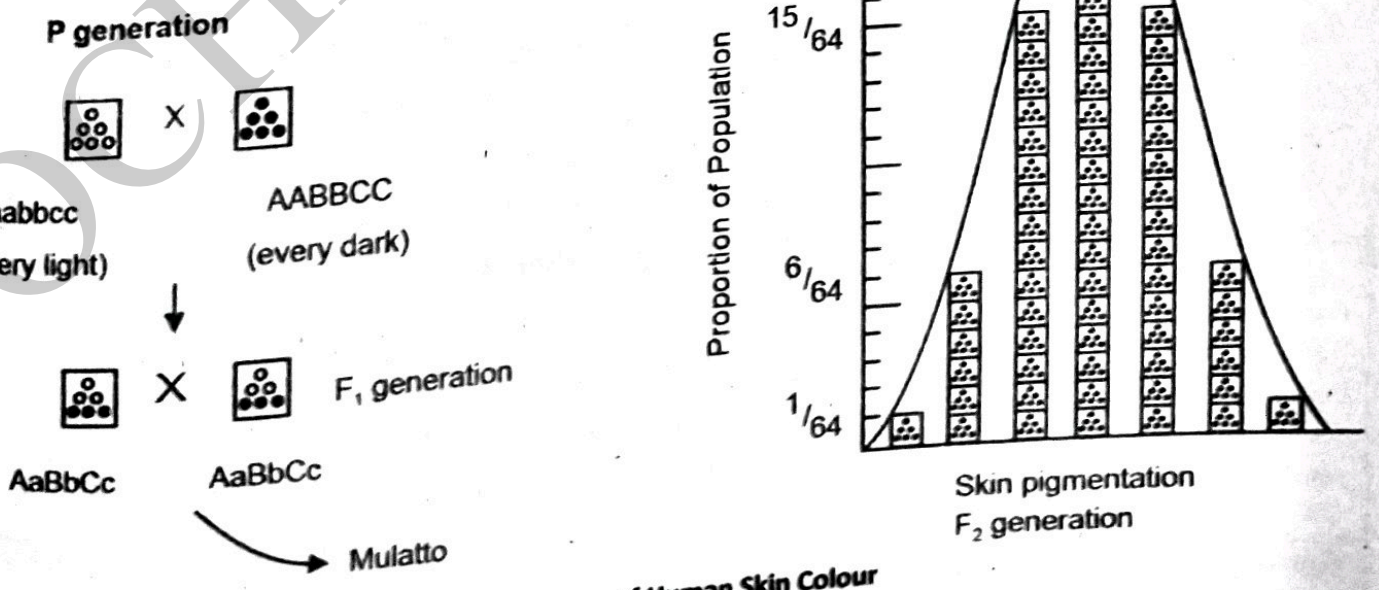



**Figure: Inheritance of Human Skin Colour**

## Examples of Polygenic Traits in Humans

### Definition of Polygenic Traits

- Polygenic traits are characteristics that are influenced by multiple genes rather than a single gene.



### Common Polygenic Traits in Humans

- Height
- Skin color variation
- Susceptibility to certain diseases, such as diabetes, cancer, and heart disease
- Intelligence
- Blood pressure levels
- Resistance to diseases
- Autism's development
- Longevity, or lifespan length
- Bipolar disorder, previously known as manic-depressive illness or manic depression

## Epistasis: Nonallelic Genetic Interactions

### Definition of Epistasis

- Epistasis is a phenomenon where the effect of one gene (epistatic gene) at a locus interferes with, or masks, the effect of another gene at a **different locus**.

### Roles of Genes in Epistasis

- The **epistatic gene** or **inhibiting gene** is the one that suppresses the effect of another gene.
- The gene whose effect is suppressed or masked is referred to as the **hypostatic gene**.

### Differentiating Epistasis from Dominance

- Epistasis should not be confused with dominance.
- Dominance involves the relationship between alleles at the **same genetic locus**, affecting how they express in traits.
- In contrast, epistasis involves interactions between different genes at **different loci**.

### Illustration of Epistasis vs. Dominance

- It is important to note that dominance affects alleles of the same gene, whereas epistasis involves the interaction across different genes.

> The expression of ABO genotypes (locus is on chromosome 9) also depends upon another gene H (locus is on chromosome 19) that encodes a particular H substance. The H substance is a precursor to the *A* and *B* antigens. For instance, the $I^B$ 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 $I^A$ allele. However, if only recessive alleles for the H substance are inherited (hh), 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 seemingly impossible phenotype result has been referred to as a Bombay phenotype because it was first described in that Indian city.

**Table – Difference between Dominance and Epistasis**

| Factor | Dominance | Epistasis |
|---|---|---|
| **Number of Allele Pairs Involved** | Involves a single pair of alleles. | Involves two pairs of alleles. |
| **Expression Suppression** | A gene suppresses the expression of its own allele. | A gene suppresses the expression of an allele of another gene. |
| **Alleles Affected** | Only the recessive allele is suppressed. | Suppresses both dominant and recessive alleles of another gene. |
| **Role of Alleles** | The effect is only due to the dominant allele. | Both dominant and recessive alleles can become epistatic. |

**Test Your Skills**

1. A couple seeks genetic counseling regarding their child's potential height, given their own heights and family history. How would you explain polygenic inheritance to them, and what factors would you consider in predicting the child's height?
2. In a genetic study of wheat grain color, what did the cross between a homozygous dark red grain plant and a homozygous white grain plant reveal about the genetic basis of color variation?
3. How does the combination of alleles from multiple genes result in a spectrum of phenotypes for polygenic traits?
4. What role do alleles A, B, and C play in determining skin color, and how might their interactions lead to variations in skin pigmentation?



## Relationship between Epistasis and Polygenic Inheritance

### Overview of Genetic Interactions

- Both epistasis and polygenic inheritance involve the interaction of more than one gene.



### Distinctive Traits in Epistasis and Polygenic Inheritance

- In epistasis, traits exhibit **discontinuous variations**. They can clearly be categorized into distinct types or classes.
- In polygenic inheritance, traits display **continuous varlations**. They result in a range of phenotypes that do not fall into distinct categories.

## Coat Color in the Labrador Retriever: An Example of Epistasis

### Basic Genetics of Coat Color

- Labrador Retrievers have three basic coat colors: yellow, black, and chocolate.
- The allele for black coat color (B) is dominant over the allele for chocolate color (b).

### Determination of Black and Chocolate Colors

- A puppy will be chocolate only if it inherits two chocolate alleles (bb) from its parents.
- If a puppy inherits at least one black allele (B), whether from one (Bb) or both parents (BB), it will have a black coat.

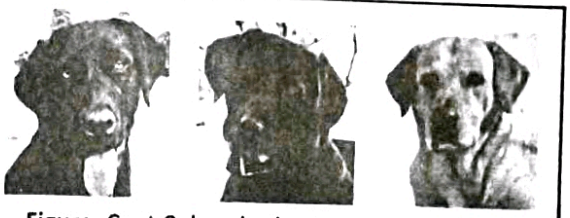


Figure: Coat Colour in the Labrador Retriever

### Role of Epistasis in Yellow Coat Color

- The gene for yellow coat color is located at a different locus in the DNA from the black versus chocolate color genes.
- To exhibit a yellow coat, a Labrador must have two recessive copies of the yellow gene (ee).
- When two yellow alleles (ee) are present, they override the effects of the black and chocolate genes, making the yellow gene epistatic.

### Breeding Implications

- Yellow Labradors (ee) will always produce yellow offspring (ee) since they can only pass on the yellow allele.
- If a black Labrador homozygous for both gene pairs (BBEE) is crossed with a yellow Labrador (bbee), all offspring will be black (BbEe) because the black gene is dominant and the yellow gene from the yellow parent is masked.
- Breeding two black Labradors heterozygous for both gene pairs (BbEe) can result in offspring with all three coat colors — black, chocolate, and yellow — in a 9:3:4 ratio.

### Genetic Problem 12.9

**When two chocolate coloured Labradors were crossed, a yellow puppy was born, what is the probability of yellow coat coloured puppy if the parents are again crossed?**

**1. Parental Genotypes**

Both parents must be Ee (heterozygous for coat color) or ee (homozygous recessive for yellow).

**2. Probability Calculation**

If both parents are Ee, each parent has a 50% chance of passing the recessive allele (e) to their offspring.
If one parent is Ee and the other is ee, each parent will pass on the recessive allele (e), resulting in a 100% chance of the offspring being ee (yellow coat).

**3. Conclusion**

If both parents are Ee, there's a 25% chance (1 in 4) of each puppy being ee (yellow coat).
If one parent is Ee and the other is ee, all puppies will be ee (yellow coat).

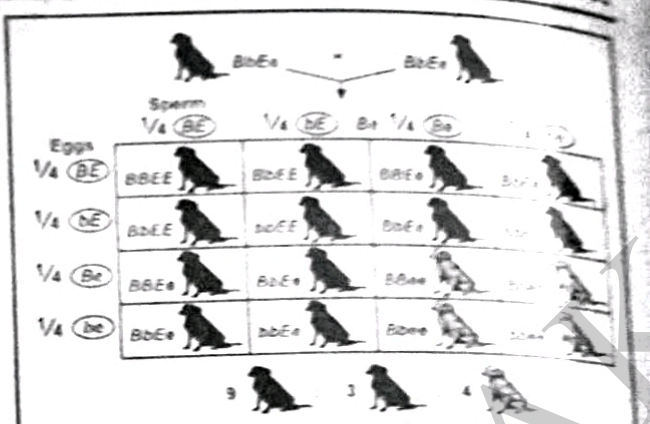



**Figure: Inheritance of Coat Colour of Labrador Retriever**



## Epistasis in Foxglove Flowers

### Genetic Interactions in Foxgloves

- In foxgloves, two genes interact to determine petal coloration, but these genes are unlinked.

**Gene Interaction for Pigment Production:**

- The wild-type allele 'd' produces light red pigment.
- The mutant allele 'D' produces dark red pigment.
- Both 'D' and 'd' alleles work on a precursor to produce a different pigment.

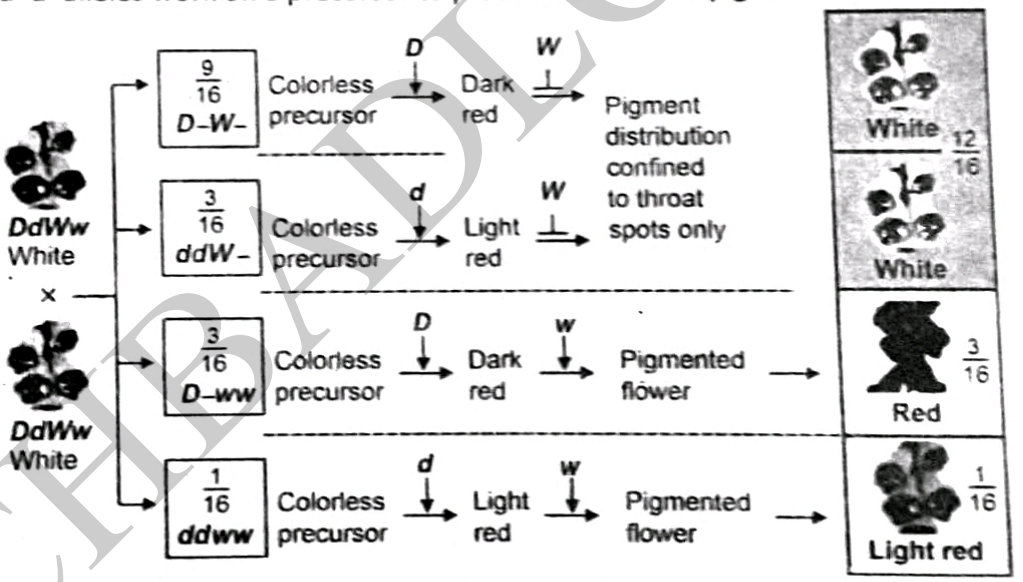



**Figure (a): Petal Colour of Foxglove Flower Exhibits Epistasis**

**Figure (b): Petal Colour of Foxglove Flower Exhibits Epistasis**

### Allelic Effects on Flower Color Distribution

- **Wild-Type Allele 'w'**: Distributes color throughout the flower.
- **Mutant Allele 'W'**: Restricts pigment to the throat of the flower, demonstrating an epistatic effect by limiting flower color.

### Phenotypic Outcomes Based on Allelic Combinations

**White Flowers:**

- 'DW' results in dark red pigment (from 'D') deposited only in throat spots.
- 'dW' results in light red pigment (from 'd') deposited only in throat spots.

**Red Flower:**

- 'Dw' produces a dark red pigment that is not restricted by 'W'.

**Light Red Flower:**

- 'dw' produces a light red pigment that is not restricted in the flower by the dominant epistatic allele 'W'.



## Inheritance of Flower Color in Sweet Pea (Lathyrus Odoratus)

### Overview of Flower Color Genetics

- Sweet pea flowers exhibit two phenotypes for color: purple (dominant) and white (recessive).

### Genetic Mechanism for Purple Color

- The purple color in sweet peas is due to the production of anthocyanin, a purple pigment.
- Anthocyanin production is controlled by two gene loci, requiring at least one dominant allele from each gene pair (A and B) to be produced.
- The dominant allele at gene "A" processes a colorless precursor into an intermediate product, which is then transformed into anthocyanin by the action of the dominant allele at gene "B".
- This interaction, where dominant alleles "A" and "B" complement each other, is known as complementary gene interaction.

### Epistasis and White Color Formation

- If either gene locus is homozygous recessive (AAbb or aaBB), the dominant alleles' expression is masked, resulting in white flowers.
- This scenario, where the recessive alleles prevent the expression of dominant alleles and lead to a different phenotype, is termed duplicate recessive epistasis.

### Genetic Cross and Offspring Phenotypes

- Crossing two white flower plants (AAbb and aaBB) results in all purple $F_1$ offspring because each parent provides one of the necessary dominant alleles.
- When these $F_1$ purple flower plants are self-fertilized, the expected Mendelian $F_2$ ratio of 9:3:3:1 is altered to 9:7, illustrating the effect of duplicate recessive epistasis in altering expected genetic ratios.

**Test Your Skills**

1. Labrador Retrievers can have coat colors such as black, chocolate, and yellow. Explain how the epistatic interaction between two genes (e.g., B and E) determines these coat colors.
2. Using a Punnett square, predict the coat color phenotypes of offspring from a cross between a black Labrador (genotype B E) and a chocolate Labrador (genotype bb E).
3. How does the concept of epistasis explain why some Labrador Retrievers may appear yellow (ee)?
4. Lathyrus odoratus, or sweet peas, exhibit flower colors ranging from purple to white. Explain how epistatic interactions between genes (e.g., A and B) control flower pigmentation.
5. In Labrador Retrievers, coat color is determined by two genes: B (black/brown pigment) and E (extension of pigment). The B gene is epistatic to the E gene. Black (B E), chocolate (bb E), and yellow (ee) are the possible phenotypes. Perform a cross between a black Labrador (B E) and a chocolate Labrador (bb E). What are the expected phenotypic ratios of their offspring?

## Multiple Choice Questions

### Polygenic Inheritance and Epistasis

1. **What does polygenic inheritance primarily describe?**
   A. Traits controlled by a single gene
   B. Random genetic mutations
   C. Traits controlled by multiple genes
   D. Traits not influenced by genetics
2. **Which type of variation do polygenic traits typically exhibit?**
   A. Discontinuous variation B. Continuous variation
   C. No variation D. Categorical variation
3. **What is an example of a polygenic trait in humans?**
   A. Blood type B. Height
   C. Eye color D. Attached earlobes
4. **How are polygenic traits encoded?**
   A. By a single gene
   B. By multiple genes with one allele
   C. By alleles of two or more different gene pairs
   D. By non-genetic factors only
5. **What is the effect of each gene in a polygenic trait?**
   A. Negligible B. Random
   C. Additive D. Counteractive
6. **What are polygenes?**
   A. Genes that mutate frequently
   B. Genes without function
   C. Genes contributing to a trait in an additive manner
   D. Genes that do not influence phenotypes
7. **What phenotype is produced by the genotype 'AABBC' in wheat grain color?**
   A. White grains
   B. Dark red grains
   C. Light pink grains
   D. Moderately dark red grains
8. **What does the homozygous genotype 'aabbcc' produce in wheat grains?**
   A. White grains B. Dark red grains
   C. Light pink grains D. Light red grains
9. **How does the environment affect wheat grain color?**
   A. It has no effect
   B. It changes genetic composition
   C. It influences pigmentation
   D. It reduces genetic diversity
10. **What primarily influences human skin color?**
    A. Single gene mutation
    B. Environmental factors only
    C. Multiple genes regulating melanin
    D. Random genetic drift
11. **What alleles promote increased melanin production?**
    A. Uppercase letters (A, B, C)
    B. Lowercase letters (a, b, c)
    C. Both uppercase and lowercase
    D. Non-genetic factors
12. **How do heterozygous combinations like 'AaBbCc' affect skin color?**
    A. Produce the darkest possible tone
    B. Result in intermediate skin tones
    C. Have no effect on skin color
    D. Lead to the lightest possible tone
13. **Which definition describes epistasis?**
    A. Interaction between identical genes
    B. Interaction where one gene masks the effect of another gene
    C. Influence of environment on gene expression
    D. Suppression of mutations by a gene
14. **What is the role of an epistatic gene?**
    A. To duplicate the effect of another gene
    B. To mutate another gene
    C. To suppress the effect of another gene
    D. To enhance the expression of another gene
15. **How is epistasis different from dominance?**
    A. Epistasis involves interactions between genes at different loci
    B. Epistasis only affects recessive alleles
    C. Dominance does not influence phenotypes
    D. Dominance involves multiple gene interactions
16. **What gene pair combination is involved in the production of anthocyanin in sweet peas?**
    A. aaBB B. AABB
    C. AaBb D. aabb
17. **What is the phenotypic outcome of the genotype 'DW' in foxglove flowers?**
    A. Dark red pigment in throat spots
    B. Uniform dark red flowers
    C. Light red pigment in throat spots
    D. No pigmentation
18. **Which scenario describes duplicate recessive epistasis?**
    A. Only dominant alleles are expressed
    B. Recessive alleles at two loci suppress the expression of dominant alleles
    C. Two dominant alleles enhance each other's effects
    D. No alleles are expressed

19. How ... affect coat color in Labrador Retrievers?
A. Changes black to chocolate
B. Has no effect
C. Masks the effect of the 'B' gene when homozygous recessive
D. Enhances pigmentation

20. What is the expected result of crossing two heterozygous black Labradors (BbEe)?
A. All black offspring
B. All chocolate offspring
C. Black, chocolate, and yellow offspring
D. Only yellow offspring

**Answer Key**

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1. C | 2. B | 3. B | 4. C | 5. C | 6. C | 7. B | 8. A | 9. C | 10. C | 11. A | 12. B | 13. B | 14. C | 15. A |
| 16. B | 17. A | 18. B | 19. C | 20. C | | | | | | | | | | |



**SLO 12.24 to 12.26** — Gene Linkage and Crossing Over

Students will be able to:

12.24 **Describe the terms gene linkage and crossing over.**

12.25 **Explain that gene linkage counters independent assortment and crossing-over modifies the progeny.**

12.26 **Suggest that linkage can be observed/evaluated only if the number of progeny is quite large.**

## GENE LINKAGE AND CROSSING OVER

- Mendel did not know the physical nature of genes. He also did not know that genes are components of chromosomes.
- The discovery of chromosomes occurred long after Mendel's experiments were concluded. Thus, Mendel's understanding of genetic mechanisms was limited.

### Gene Linkage

**Basic Concept of Gene Linkage**

- Each chromosome contains hundreds to thousands of genes.
- Genes located on the same chromosome that are **inherited together** are referred to as **linked genes**.
- The phenomenon where multiple genes on the same chromosome are inherited together is called **gene linkage**.

**Types of Gene Linkage**

**Autosomal Linkage**

- This occurs when genes are linked on autosomes, the non-sex chromosomes.

**Sex Linkage**

- This refers to genes linked on sex chromosomes.

**Linkage Groups**

- Linked genes on the same homologous pair of chromosomes form a linkage group.
- The number of linkage groups in an organism equals the number of homologous pairs of chromosomes.

**Impact on Genetic Inheritance**

- Linked genes tend to be inherited together and generally do not undergo recombination.
- Because of this, linked genes do not assort independently, leading to deviations from the Mendelian ratio of independent assortment.

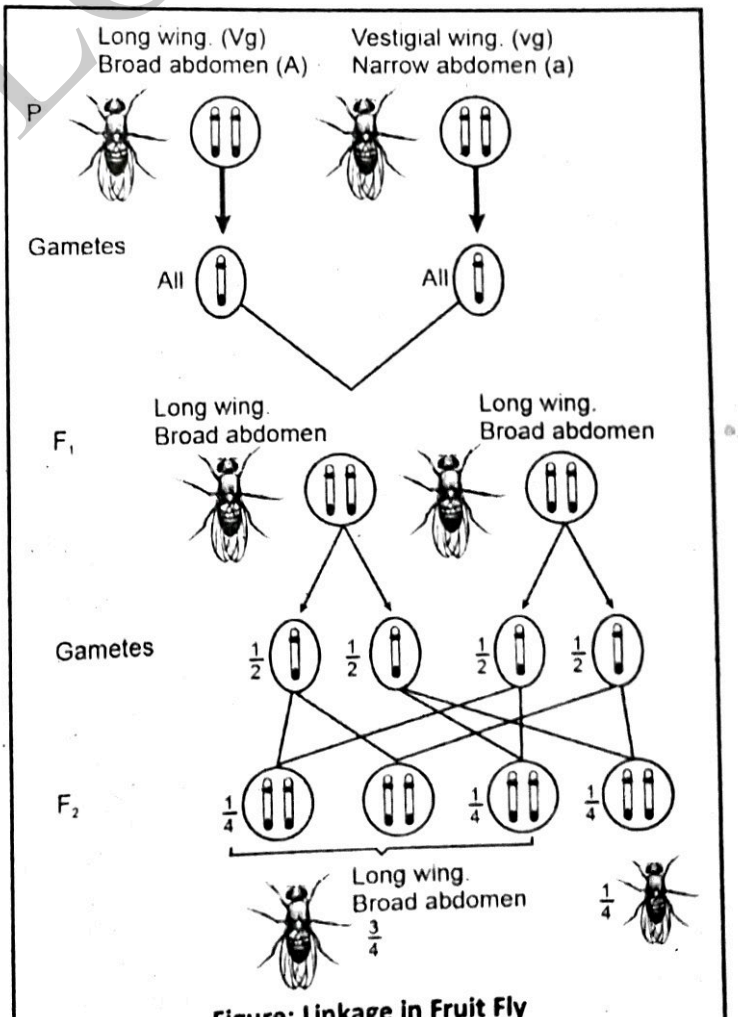



**Figure: Linkage in Fruit Fly**

## Detection of Gene Linkage

### Method of Detecting Gene Linkage

- Gene linkage can be detected by conducting a test cross involving two gene pairs, known as a dihybrid test cross.
- In this cross, an individual heterozygous for two traits ($F_1$) is backcrossed with its homozygous recessive parent ($P_2$).

### Analyzing Outcomes of a Test Cross

- If all four phenotypic combinations (both parental and recombinant types) are produced in an equal 1:1:1:1 ratio, it suggests that there is no linkage between the genes.
- A deviation from this ratio, characterized by a predominance of parental types and fewer recombinant types, indicates incomplete linkage.
- The production of only parental types suggests complete linkage.

### Impact of Complete or Tight Linkage

- In cases of complete or tight linkage, recombinant types are not produced, which disturbs the typical 9:3:3:1 ratio observed in independent assortment, resulting instead in a 3:1 ratio of parental combinations only.

### Example of Linkage Study

- T. H. Morgan's experiments with Drosophila provide a clear example of how linkage between genes affects the inheritance of different traits.

## Morgan's Experiment on Drosophila Melanogaster

### Overview of Traits Studied

- T. H. Morgan investigated approximately 85 pairs of contrasting traits in the fruit fly, Drosophila melanogaster.
- Among these traits were **wing length** and **abdomen width**.

### Genetic Dominance in Traits

- The allele for long wings (Vg) is dominant over the allele for short or vestigial wings (vg).
- Similarly, the allele for a broad abdomen (A) is dominant over the allele for a narrow abdomen (a)

### Initial Cross and $F_1$ Generation

- Morgan crossed a fruit fly with long wings and a broad abdomen with another fly that had vestigial wings and a narrow abdomen.
- All $F_1$ offspring exhibited the dominant traits: long wings and broad abdomens.

### $F_2$ Generation Observations

- When two $F_1$ flies were mated, about 3/4 of the $F_2$ offspring displayed long wings and broad abdomens.
- Nearly all the remaining 1/4 of the $F_2$ flies exhibited the recessive traits: vestigial wings and a narrow abdomen.

## Gene Linkage versus Independent Assortment in Morgan's Experiment

### Expectation from Independent Assortment

- Based on Mendel's law of independent assortment, which predicts a 9:3:3:1 phenotypic ratio in dihybrid crosses, Morgan expected a similar distribution in his experiments with Drosophila.

### Observations Deviating from Expectations

- Morgan observed a different pattern in the $F_2$ generation: most offspring either had both dominant traits or both recessive traits, resulting in an approximately 3:1 ratio rather than the expected 9:3:3:1.

### Conclusion on Gene Linkage

- From his data, Morgan concluded that the genes for abdomen width and wing length were located on the same chromosome.
- This close proximity meant that these genes did not assort independently during meiosis.

### Impact of Linkage on Genetic Assortment

- Because the genes were linked, they were inherited together, reducing the occurrence of recombinant types.
- This linkage altered the expected Mendelian ratio of independent assortment from 9:3:3:1 to 3:1, demonstrating the effect of gene linkage on phenotypic outcomes in genetic crosses.

## Crossing-Over: Modifying Progeny through Genetic Recombination

### Process of Crossing-Over



- Crossing-over is the process responsible for the recombination of linked genes.
- It involves the exchange of segments between maternal and paternal chromatids during prophase of meiosis I, while homologous chromosomes are paired.

### Impact on Genetic Variation

- The recombinant chromatids formed as a result of crossing-over may carry new combinations of alleles.
- This results in the production of a diverse array of gametes, each potentially carrying different genetic information.

### Contribution to Genetic Variability

- Crossing-over contributes to genetic variability in sexually reproducing organisms.
- It enables the exchange of genetic material between maternal and paternal chromosomes.

### Effect on Offspring

- The genetic recombination from crossing-over results in offspring that possess different combinations of genes than their parents.
- Consequently, crossing-over plays a crucial role in modifying the genetic outcomes in progeny, enhancing the diversity within a species.

**Test Your Skills**

1. How does crossing-over contribute to genetic diversity?
2. Discuss the difference between the expected 9:3:3:1 ratio of independent assortment and the 3:1 ratio observed in linkage situations.
3. How did Morgan conclude that certain traits in Drosophila were linked?
4. How do linked genes violate Mendel's law of independent assortment?

## Multiple Choice Questions

### Gene Linkage and Crossing Over

1. **What was Mendel unaware of during his genetic studies?**
   A. Physical nature of genes
   B. Role of proteins in traits
   C. Effects of the environment
   D. Mechanisms of natural selection
2. **What are linked genes?**
   A. Genes that mutate together
   B. Genes that express the same trait
   C. Genes that skip generations
   D. Genes on the same chromosome inherited together
3. **What defines autosomal linkage?**
   A. Linkage affecting only males
   B. Linkage that skips generations
   C. Linkage on non-sex chromosomes
   D. Linkage causing mutations
4. **What results from gene linkage?**
   A. Genes assort independently
   B. Genes tend not to undergo recombination
   C. Increased genetic mutations
   D. Decreased genetic diversity
5. **How can gene linkage be detected?**
   A. Pedigree analysis B. Mutation testing
   C. Dihybrid test cross D. Environmental study
6. **What suggests complete linkage in a test cross?**
   A. An equal ratio of all phenotypic combinations
   B. Only parental types are produced
   C. A continuous variation in traits
   D. Recombinant types outnumber parental types
7. **What was a key finding from Morgan's experiments with Drosophila?**
   A. Genes assort randomly
   B. Traits are influenced by environmental factors
   C. Linked genes do not assort independently
   D. All traits are controlled by a single gene

8. **Which traits did Morgan study in Drosophila melanogaster?**
   A. Eye color and body size
   B. Wing length and abdomen width
   C. Leg length and antenna size
   D. Reproductive and digestive traits
9. **What is a common result of gene linkage?**
   A. Increased mutation rate
   B. Deviation from Mendelian ratio of independent assortment
   C. Decreased genetic diversity
   D. Stabilization of trait expression
10. **How does crossing-over affect offspring?**
   A. Reduces genetic variation
   B. Creates new combinations of genes
   C. Stabilizes genetic traits
   D. Eliminates recessive alleles
11. **What is the significance of crossing-over during meiosis?**
   A. It prevents gene linkage
   B. It leads to genetic recombination
   C. It increases chromosome number
   D. It decreases genetic diversity
12. **What did Morgan's experiments demonstrate about gene linkage?**
   A. Linked genes are inherited together more frequently than not
   B. Genes on different chromosomes are linked
   C. Environmental factors can break gene linkage
   D. Linkage can occur without genetic inheritance
13. **How does the 3:1 ratio observed in Morgan's experiments relate to linkage?**
   A. It confirms independent assortment
   B. It is typical for unlinked genes
   C. It indicates linked genes are not assorting independently
   D. It suggests multiple alleles are involved
14. **Which type of gene linkage involves genes on the sex chromosomes?**
   A. Autosomal linkage B. Chromosomal linkage
   C. Sex linkage D. Reciprocal linkage
15. **What does a deviation from a 1:1:1:1 ratio in offspring suggest?**
   A. Independent assortment
   B. Mutation occurrence
   C. Incomplete linkage
   D. Environmental influence
16. **What happens during the process of crossing-over?**
   A. Chromosomes duplicate
   B. Segments between chromatids are exchanged
   C. Genes are deleted from chromosomes
   D. Chromosomes are entirely swapped
17. **What is the effect of complete linkage?**
   A. Production of only recombinant types
   B. Random assortment of genes
   C. No recombinant types produced
   D. Independent assortment is confirmed
18. **What is indicated by the production of only parental types in a test cross?**
   A. Independent assortment
   B. Complete linkage
   C. No linkage
   D. Environmental influence
19. **How is the number of linkage groups determined in an organism?**
   A. By the number of genes
   B. By the length of chromosomes
   C. By the strength of gene expression
   D. By the number of homologous pairs of chromosomes
20. **What does the presence of both parental and recombinant types in similar ratios indicate?**
   A. Complete linkage B. No linkage
   C. Autosomal linkage D. Sex linkage

**Answer Key**

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1. A | 2. D | 3. C | 4. B | 5. C | 6. B | 7. C | 8. B | 9. B | 10. B | 11. B | 12. A | 13. C | 14. C | 15 C |
| 16. B | 17. C | 18. B | 19. D | 20. B | | | | | | | | | | |

**SLO 12.27 to 12.29 — Sex Determination**

**Students will be able to:**

**12.27 Explain the - mechanism of sex determination in mammals.**
**12.26 Identify male and female individuals from the karyotype of man.**
**12.27 Solve the genetic problems related to $XX - XY$, sex determination.**

# SEX DETERMINATION - CHROMOSOMAL BASIS

The difference between male and female in most animals and plants is genetically determined by their karyotype.

A karyotype is the complete set of chromosomes in an organism.



## Chromosomes in Drosophila

Drosophila, commonly known as fruit flies, have a total of eight chromosomes, arranged in four pairs.

Three of these pairs are autosomes, which are identical in both males and females.

The fourth pair consists of sex chromosomes, which differ between males and females:

- Females have two identical sex chromosomes, both called XX.
- Males have one X chromosome, similar to those in females, and one Y chromosome, which is hook-shaped.

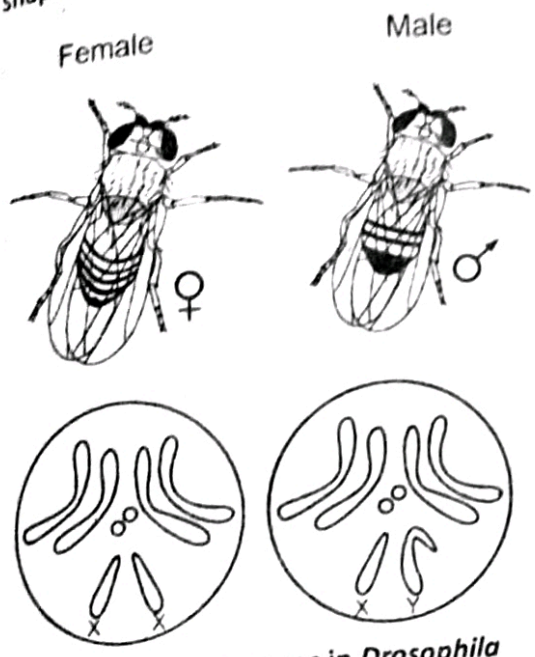



**Figure: Chromosomes in *Drosophila***

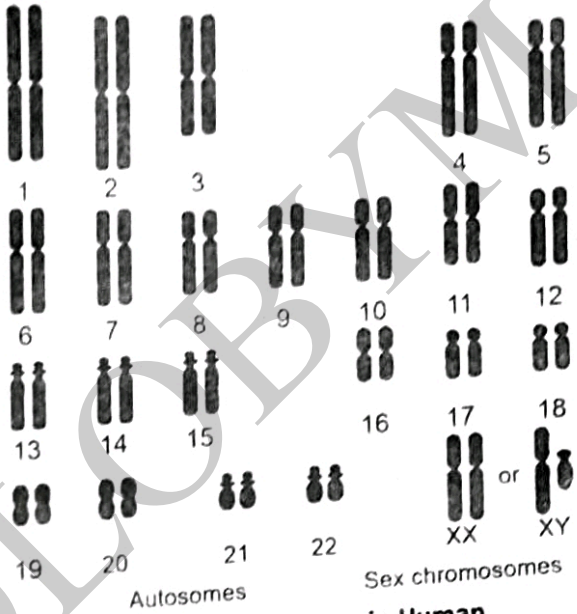



**Figure: Chromosomes in Human**

## Chromosomes in Humans

Humans have a total of 46 chromosomes. These are organized into 23 pairs.

22 these pairs are autosomes. They are the same in both males and females.

The 23rd pair consists of **sex chromosomes**:

- Females have **two X** chromosomes (XX).
- Males have **one X** chromosome and **one Y** chromosome (XY).

All egg cells produced by a woman contain one X chromosome.

Sperm cells from a man contain either an X or a Y chromosome.

The sex of a child is determined by the type of sex chromosome in the sperm that fertilizes the egg:

- An X chromosome from the sperm results in a female child (XX).
- A Y chromosome from the sperm results in a male child (XY).

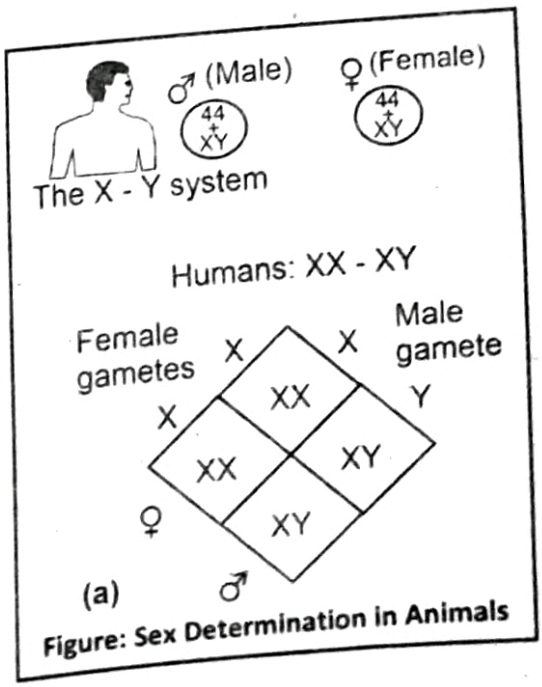



**Figure: Sex Determination in Animals**

## Patterns of Sex Determination

### X-XX Type

This common sex determination system is observed in humans, Drosophila, and many other organisms.

**Males (XY):** They are heterogametic, producing two types of sex-determining sperm. Half of the sperm carry an X chromosome, and the other half carry a Y chromosome. The probability of producing each type of sperm is equal.

- **Females (XX):** They are homogametic, producing only one type of egg, each containing an X chromosome.
- **Determining the Sex of Offspring:** The sex of the offspring is determined by the type of sperm that fertilizes the egg.
  - If an X-carrying sperm fertilizes the egg, the resulting zygote will be XX, producing a female offspring.
  - If a Y-carrying sperm fertilizes the egg, the resulting zygote will be XY, producing a male offspring.
- **Sex Ratio:** The expected ratio of male to female offspring is 1:1, indicating equal chances for the birth of a son or daughter.



**Science Titbits**

Investigators have found that the Y chromosome has an SRY gene (sexdetermining region Y gene). When this region in a gene is lacking from the Y chromosome, the individual is a female even though the chromosomal inheritance is XY. This gene is Y-linked because it is found only on the Y chromosome


**Test Your Skills**

1. Compare the XY-XX sex determination system in humans with that in Drosophila
2. Tell the probability of producing X- and Y-carrying sperm in males.
3. What similarities and differences exist in the mechanisms of sex determination between these two species?
4. How do males and females differ in their production of gametes (sperm and egg) in the XY-XX system?

### Genetic Problem 12.10

**What are the chances for a husband and wife having either a boy or girl 50:50?**

**Ans.** The chances for a husband and wife having either a boy or girl are 50:50 due to the equal probability of the father's sperm carrying either an X (resulting in a girl) or a Y chromosome (resulting in a boy), paired with the mother's X chromosome. This leads to an equal likelihood of conceiving either gender.

### Genetic Problem 12.11

**A woman produced four daughters in successive pregnancy. What are the prospects of her getting a son in the fifth pregnancy if it occurs?**

**Ans.** The likelihood of a woman having a son in her fifth pregnancy remains 50%. Each pregnancy is an independent event determined by the random combination of the father's sperm carrying either an X (girl) or Y chromosome (boy), maintaining an equal chance for either gender.

### Genetic Problem 12.12

**If both parents are $A^{+ve}$ and have an $O^{-ve}$ child. Calculate the probability of $A^{+ve}$ daughter and $A^{+ve}$ son in next pregnancy?**

**Ans.** If both parents are $A^{+ve}$ (Rh-positive) and have an $O^{-ve}$ ) child, the probability in the next pregnancy is:

– 50% chance of an $A^{+ve}$ (Rh-positive) daughter.

– 50% chance of an $A^{+ve}$ (Rh-positive) son, since the Rh factor inheritance is independent in each pregnancy.

## Multiple Choice Questions

### Sex Determination

1. **What is a karyotype?**
   A. A type of genetic mutation
   B. A specialized chromosome structure
   C. A complete set of chromosomes in an organism
   D. A method to alter genetic information
2. **How many chromosomes do humans have?**
   A. 8 B. 22
   C. 46 D. 23
3. **How many chromosome pairs are autosomes in humans?**
   A. 22 B. 23
   C. 21 D. 20
4. **What characterizes the sex chromosomes in human females?**
   A. XY B. XYY
   C. XX D. X
5. **How many types of sperm cells do human males produce concerning the sex chromosomes?**
   A. One type B. Two types
   C. Three types D. Four types
6. **Which chromosomes do female egg cells contain?**
   A. Y B. XY
   C. X D. XX
7. **What results in a female child in XY-XX sex determination?**
   A. An X-carrying sperm fertilizes the egg
   B. A Y-carrying sperm fertilizes the egg
   C. Both X and Y carrying sperm fertilize the egg
   D. No sperm fertilizes the egg
8. **How many chromosomes are in Drosophila?**
   A. 8 B. 23
   C. 46 D. 22
9. **What is true about the sex chromosomes in male Drosophila?**
   A. XX B. YY
   C. XY D. XXY
10. **In XY-XX sex determination, which sex is heterogametic?**
    A. Males B. Females
    C. Both are heterogametic
    D. Neither is heterogametic
11. **What is the expected ratio of male to female offspring in XY-XX sex determination?**
    A. 2:1 B. 1:2
    C. 1:1 D. 3:1
12. **Which chromosomes determine gender?**
    A. Y-chromosomes B. XX-chromosomes
    C. Sex-chromosomes D. None of these
13. **What are the chances of a couple having a boy or a girl in each pregnancy?**
    A. 50:50 B. 75:25
    C. 25:75 D. 60:40
14. **Each pregnancy is considered an independent event in terms of predicting the sex of the offspring. What does this mean?**
    A. Each child will be the same sex
    B. The sex of one child affects the sex of the next
    C. The probability of having a boy or girl resets with each pregnancy
    D. All children will be of different sexes
15. **If both parents have positive Rh factors but had an O-negative child, what is the chance of having an A-positive child in the next pregnancy?**
    A. 25% B. 75%
    C. 50% D. 100%
16. **How do autosomal and sex linkage differ?**
    A. Autosomal linkage occurs on autosomes, sex linkage on sex chromosomes
    B. There is no difference
    C. Autosomal linkage does not affect inheritance
    D. Sex linkage only occurs in males
17. **What aspect of sperm determines the sex of the offspring in XY-XX systems?**
    A. Size of the sperm B. Quantity of sperm
    C. Type of sex chromosome in the sperm
    D. Speed of the sperm
18. **What was the initial observation in Morgan's Drosophila cross concerning offspring traits?**
    A. All F1 had recessive traits
    B. All F1 exhibited dominant traits
    C. F1 showed new mutations
    D. F1 were infertile
19. **What pattern of inheritance did Morgan observe that differed from expected Mendelian ratios?**
    A. Predominance of either all dominant or all recessive traits
    B. An equal mix of dominant and recessive traits
    C. Disappearance of dominant traits
    D. Only new mutations appeared
20. **What is the role of crossing-over in genetic recombination?**
    A. Prevents recombination
    B. Facilitates the exchange of genetic material between chromatids
    C. Stabilizes the chromosome structure
    D. Reduces the number of chromosomes

**Answer Key**

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1. C | 2. C | 3. A | 4. C | 5. B | 6. C | 7. A | 8. A | 9. C | 10. A | 11. C | 12. B | 13. A | 14. C | 15. C |
| 16. A | 17. C | 18. B | 19. A | 20. B | | | | | | | | | | |

**SLO 12.30 to 12.35** — Sex Linkage

Students will be able to:

12.30 Describe the concept of sex-linkage.
12.31 Explain the inheritance of sex-linked traits (eye-colour) in Drosophila.
12.32 Describe the sex-linked inheritance of male characters due to Y-chromosome and the effect of Hollandric genes.
12.33 Describe the X-linked disorders with reference to the patterns of inheritance.
12.34 Name some of the sex-linked disorders of man (Red green colour blindness, Hemophilia).
12.35 Explain the techniques employed for embryonic screening e.g., Amniocentesis and chronic villus sampling.

# SEX LINKAGE

- Sex linkage are the traits that are controlled by alleles located on the sex chromosomes.

### Types of Sex-Linked Traits

- **X-linked Traits**: These traits are controlled by alleles that are located only on the X chromosome.
- **Y-linked Traits**: These traits are controlled by alleles that are located only on the Y chromosome.

### X-Y Linked or Pseudo Autosomal Traits

- Some traits have genes with alleles on both the X and Y chromosomes. These are referred to as X-Y linked or pseudo autosomal traits.
- Unlike other sex-linked traits, pseudo autosomal traits inherit like autosomal traits, where genes are located on non-sex chromosomes.
- Very few alleles are found on the Y chromosome, limiting the number of Y-linked traits.

## Sex Linkage in Drosophila: The Discovery of X-Linked Inheritance

- Thomas Hunt Morgan conducted experiments on Drosophila melanogaster to study sex linkage.
- Wild-type Drosophila typically have bright red eyes, while mutations can result in white eyes.
- Morgan focused on eye color inheritance.

### Initial Observations and Crosses

- Morgan's initial cross involved a white-eyed male and a red-eyed female, producing 1237 red-eyed F1 progeny, indicating that red-eye color is dominant.
- In the F2 generation, obtained by crossing F1 individuals among themselves, there were 3470 red-eyed flies and 782 white-eyed flies, with all white-eyed flies being male.

### Deviations from Mendelian Ratios

- Morgan observed two major deviations from Mendelian inheritance:
  1. The F2 generation showed a phenotypic ratio that slightly deviated from the expected 3:1 ratio of a monohybrid cross.
  2. All white-eyed flies in the F2 generation were male, suggesting a sex-linked trait.

### Genetic Explanation

- Morgan noted that the eye color gene is located on the X chromosome (denoted as XR for red and Xr for white), while the Y chromosome does not carry this gene.
- Females are XX and can be either red-eyed (XRXR or XRXr) or white-eyed (XrXr), while males are XY, with red eyes (XRY) or white eyes (XrY).

### Further Testing and Confirmation

- Morgan conducted a test cross of red-eyed F2 females with the original white-eyed male, resulting in offspring of all possible combinations, which included 88 white-eyed females, confirming that the trait could appear in females and was not lethal.

- A subsequent mating of a white-eyed female with a red-eyed male resulted in all-female offspring having red eyes and all male offspring having white eyes, further confirming the X-linked inheritance.

### Morgan's Conclusion

- Morgan concluded that the white-eye trait was carried on the X chromosome and that the Y chromosome did not contribute to this trait.
- His findings were consistent with Mendelian segregation but adjusted for the complications introduced by sex-linked inheritance, thereby solidifying the concept of X-linked recessive traits.

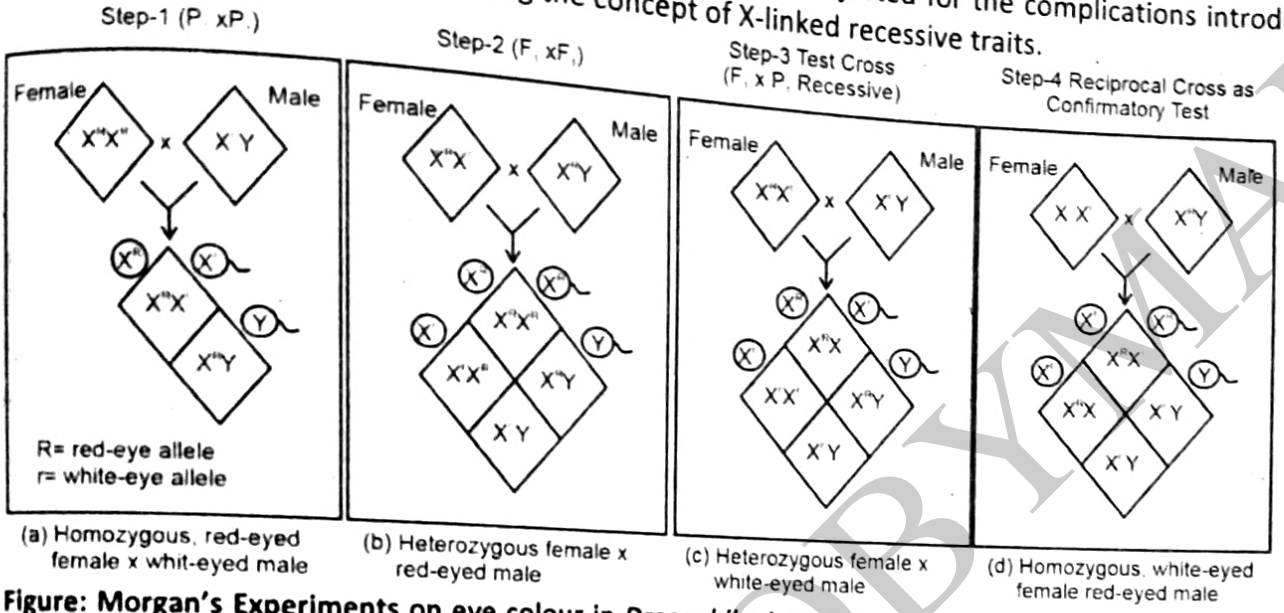



**Figure: Morgan's Experiments on eye colour in *Drosophila*. Letter R represents the dominant allele for red eye and letter r represents the recessive white eye. Because the alleles are carrier on the X chromosome, these are shown as superscript to the letter X. Thus the red eyed male fruit flies have genotype $X^RY$, white eyed males are $X^rY$. The Y chromosome does not have a gene locus for eye colours therefore; the phenotype of the male results entirely from this single X linked genes. In the female $X^RX^R$ and $X^RX^r$ have red eyes and $X^rX^r$ flies have white eyes.**



## Sex-Linkage in Humans

### Overview of X-Linked Traits

- Humans exhibit various X-linked traits, with some being recessive (e.g., hemophilia, color blindness) and others being dominant (e.g., hypophosphatemic rickets).
- **X-Linked Dominant Traits:** These are determined by a gene located on the X chromosome that manifests in both males and females if present.
- **X-Linked Recessive Traits:** These are also located on the X chromosome but typically only express in males or carrier females without showing symptoms.

### Challenges in Human Genetic Research

- Unlike experimental mating possible in other species, human genetic studies rely on pedigree analysis due to ethical restrictions.
- Human geneticists utilize pedigrees to trace the inheritance of traits, following agreed-upon standards and symbols for clarity and consistency.

Figure: Hairy Pinna

### Patterns of Inheritance

#### X-Linked Recessive Inheritance

- Carrier Females: Females with one X-linked recessive mutation generally do not show symptoms but can pass the mutation to offspring.
- Affected Males: Males with the mutation will show symptoms as they have only one X chromosome.
- Offspring of Carrier Females: Each child has a 50% chance of inheriting the mutation.

- Daughters of Affected Fathers: All will be carriers.
- Sons of Affected Fathers: Will not be affected as they do not inherit their father's X chromosome.

**X-Linked Dominant Inheritance**

- Children of Affected Mothers: Each has a 50% chance of inheriting the mutation.
- Daughters of Affected Fathers: All will be affected.
- Sons of Affected Fathers: Will not be affected.



**Y-Linked Inheritance (Holandric Traits)**

- Y-linked traits are inherited strictly from father to son as only males possess a Y chromosome.
- Phenotypic expression is exclusively male due to the location of these genes on the non-homologous region of the Y chromosome.
- Examples include hypertrichosis, porcupine man syndrome, and webbing of toes.

## Applications of Genetic Research

- Pedigree analysis remains a powerful tool in understanding the transmission of these traits across generations, aiding in the prediction and management of genetic disorders.

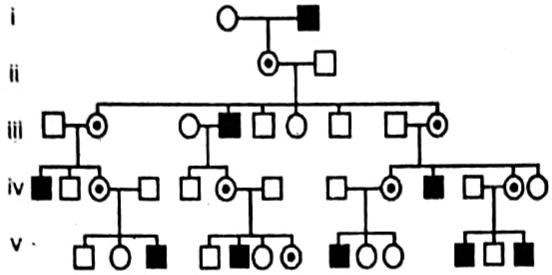



**Figure: Pedigree of recessive X Linked**

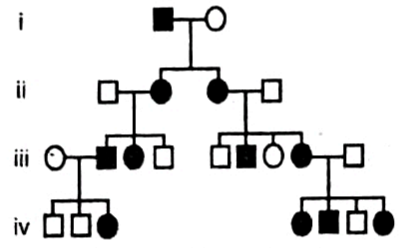



**Figure: Pedigree of Dominant X Linked**

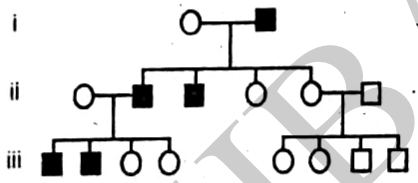
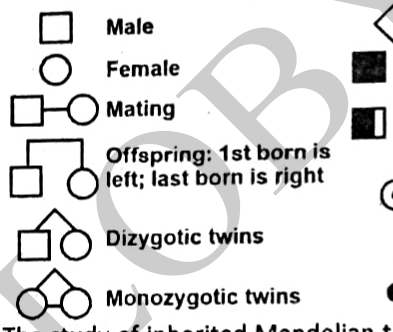



**Figure: Pedigree of Y-Linked Trait**

**Science Titbits**




The study of inherited Mendelian traits in humans must rely on observations made while working with individual families. Classical cross fertilization breeding experiments as performed by Mendel are not allowed in humans. Human geneticists are not allowed to selectively breed for the traits they wish to study. One of most powerful tools in human genetic studies is pedigree analysis. When human geneticists first began to publish family studies, they used a variety of symbols and conventions. Now there are agreed upon standards for the construction of pedigrees.

**Test Your Skills**

1. How does understanding sex linkage and inheritance patterns help in predicting and managing genetic disorders in humans?
2. Why X-linked recessive traits are more commonly observed in males than in females?

# Sex-Linked Disorders in Humans

## Overview of Sex-Linked Disorders

- Humans can exhibit disorders caused by mutations in sex chromosomes, similar to how Drosophila may have white eye color due to such mutations.

## Genetics of Hemophilia

- Hemophilia is a notable X-linked recessive disorder where affected individuals have impaired blood clotting due to a deficiency or absence of certain clotting factors.

- It is classified into three types: A, B, and C.

### Hemophilia A and B

- **Inheritance Pattern**: Hemophilia A and B are inherited as X-linked recessive traits.
- **Gender Prevalence**: Primarily affects males more than females.
  - Males with one affected X chromosome ($X^hY$) will express the disorder.
  - Females must have two affected X chromosomes ($X^hX^h$) to express the disorder, making it rarer in females.
- **Transmission**: The gene can be transmitted from an affected maternal grandfather through a carrier daughter to her sons. It is not directly passed from father to son because a father passes his Y chromosome, not his X, to his sons.

### Hemophilia C

- **Inheritance Pattern**: Unlike A and B, Hemophilia C is autosomal recessive, meaning it is not linked to the sex chromosomes but to autosomes (non-sex chromosomes).
- **Gender Prevalence**: Affects males and females equally, as it depends on inheriting two recessive alleles, one from each parent.
- **Transmission**: Can be passed down from either parent to any child regardless of gender.

### Clinical Implications

- **Severity**: Hemophilia can be severe, with affected individuals potentially bleeding to death from minor injuries due to the lack of proper blood clotting mechanisms.
- **Management**: It requires careful management, including treatment with clotting factor concentrates to prevent or control bleeding episodes.



### Understanding Gene Notation in Hemophilia

- **The gene for Normal Clotting:** Denoted as H.
- **The gene for Hemophilia A:** Denoted as h.
- This notation helps in understanding genetic transmission patterns and predicting the likelihood of the disorder appearing in offspring.

**Table: Haemophilia**

| No. | Types | Cause | Percentage | Inheritance |
|---|---|---|---|---|
| 1. | **Hemophilia A** | Abnormality of blood clotting factor VIII | 80% | Recessive X linked |
| 2. | **Haemophilia B** | Disturbance in blood clotting factor IX | 20% | Recessive X linked |
| 3. | **Haemophia C** | Reduction in blood clotting factor XI | Less than 1% | Recessive autosomal |

### Genetic Problem 12.13:

**Can two normal parents have a haemophiliac child? Work out the probability.**

**Solution:**

### Haemophilia Genetics

- Haemophilia is an X-linked recessive disorder.
- Males have one $X$ and one $Y$ chromosome (XY).
- Females have two X chromosomes (XX).

### Parent Genotypes

- Normal male: $XY$
- Normal female: $XX$ or $X^HX$ (carrier, where $X^H$ represents the X chromosome with the haemophilia allele)

A normal male ($XY$) cannot be a carrier because he has only one X chromosome. A normal female ($XX$) can be a carrier if she has one normal allele and one haemophilia allele ($X^HX$).

### Possible Scenarios

1. **Normal Male $XY$ and Normal Female :**
   - No haemophilia alleles are present. They cannot have a haemophiliac child.

2. Normal Male $XY$ and Carrier Female $X^HX$ :
- The carrier female has a 50% chance of passing the $X^H$ (haemophilia) allele to her offspring

**Offspring Genotypes**

- Daughters:
- Inherit one $X$ from each parent.
- $XX$ (normal) or $X^HX$ (carrier)
- Sons:
- Inherit $X$ from mother and $Y$ from father.
- $XY$ (normal) or $X^HY$ (haemophiliac)



**Probabilities**

- For a son to be haemophiliac ($X^HY$) :
- Probability of mother passing $X^H$: 50% or 0.5
- Probability of father passing Y: 100% or 1
- Combined probability: $0.5 \times 1 = 0.5$ or 50%
- For a daughter to be haemophiliac ($X^HX^H$) :
- Since the father is normal ($XY$), he cannot pass $X^H$.
- Therefore, the daughter cannot be haemophiliac.

## Justification of Hemophilia Treatments

### Treatments for Hemophilia A

1. **Use of Desmopressin**
   - **Effectiveness**: Desmopressin is effective for mild cases of Hemophilia A as it stimulates the body to produce more clotting factor VIII, which is deficient in these patients.
   - **Mechanism**: Administered typically via injection, desmopressin works by temporarily boosting the levels of factor VIII, thereby improving the clotting process in patients who still can produce some factor VIII naturally.
2. **Use of Octocogalfa**
   - **Effectiveness**: For more severe cases of Hemophilia A, octocogalfa, a synthetic version of clotting factor VIII, is used. This treatment is crucial as it directly replaces the missing or defective factor VIII, essential for effective blood clotting.
   - **Mechanism**: Octocogalfa is a genetically engineered protein that provides a consistent supply of factor VIII, thus helping to manage and prevent bleeding episodes effectively.

### Treatment for Hemophilia B

**Use of Nonacogalfa**

- **Effectiveness**: Nonacogalfa, an engineered version of clotting factor IX, is used to treat Hemophilia B, which results from a deficiency in this specific factor.
- **Mechanism**: Similar to treatments for Hemophilia A, nonacogalfa provides the necessary clotting factor IX that these patients lack, helping to control and prevent bleeding.

### Understanding the Clotting Process

**Clotting Cascade**

- Upon injury, a cascade of chemical reactions occurs, involving multiple clotting factors that lead to the formation of a blood clot. This cascade starts with the activation of platelets and damaged tissue releasing chemicals, which interact with clotting factors in the plasma.

**Final Steps**

- The cascade culminates in converting fibrinogen (factor I) into fibrin, which forms a mesh that traps blood cells and solidifies into a clot. This process involves up to 13 known clotting factors, each identified by Roman numerals I to XIII.

### Importance of Treatment

**Prevention of Bleeding**

- These treatments are essential in preventing excessive bleeding and managing bleeding episodes by ensuring that the necessary clotting factors are available in sufficient amounts.

**Quality of Life**

- Effective management of hemophilia with these treatments allows individuals to lead more normal lives, reducing the risk of complications from uncontrolled bleeding.

The treatments for hemophilia, through replacement therapy or stimulation of natural factor production, are important in managing the disorder. They are designed to address the specific deficiencies in clotting factors that characterize different types of hemophilia, thereby directly impacting the clotting cascade and enhancing the body's ability to form clots.



## Genetics of Colour-blindness

### Normal Trichromatic Colour Vision

- Normal trichromatic colour vision is based on three different kinds of **cones** in the retina.
- Each cone is sensitive to one of the three primary colours: **red**, **green**, or **blue**.
- Each type of cone cell has specific light-absorbing proteins called **opsins**.

### Opsin Genes Location

- The genes for red and green opsins are located on the X chromosome.
- The gene for blue opsin is located on autosome 7.

### Mutations and Colour-blindness

- Mutations in **opsin genes** can cause colour-blindness. It results in conditions like dichromacy and monochromacy.

### Dichromacy

- A dichromate can perceive two primary colours but is unable to perceive the one whose opsin is missing due to mutation.
  - **Protanopia**: Red blindness
  - **Deuteranopia**: Green blindness
  - **Tritanopia**: Blue blindness

### Monochromacy

- A monochromate can perceive only one colour. Monochromacy represents true colour-blindness.

**Blue Cone Monochromacy**

- An X-linked recessive trait where both red and green cone cells are absent, leading to red-green colour-blindness. It is a common hereditary disease.

### Inheritance Pattern of Blue Cone Monochromacy

- Like any sex-linked recessive trait, blue cone monochromacy follows a **zigzag pattern** of inheritance:
- It passes from the maternal grandfather through a carrier daughter to a grandson.
- It never passes directly from father to son.

### Genetic Problem:

**When a carrier women get married with normal man, what will be the probably colour blind son in their children?**

### Given Information

- Carrier woman genotype: $X^cX$ (where $X^c$ represents the X chromosome carrying the color blindness allele)
- Normal man genotype: XY

### Possible Offspring Genotypes

1. **Daughters:**
   - Can inherit one $X$ from each parent.
   - Possible genotypes: $XX$ (normal) or $X^cX$ (carrier)
2. **Sons:**
   - Can inherit $X$ from mother and $Y$ from father.
   - Possible genotypes: $XY$ (normal) or $X^cY$ (color blind)

**Probabilities**

**We need to calculate the probability of having a color blind son.**

1. **Probability of mother passing $X^c$ to a child:**
   - 50% (since the mother has one normal $X$ and one $X^c$ )
2. **Probability of having a son:**
   - 50% (since each child has an equal chance of being male or female)

### Combined Probability

- Probability of having a color-blind son:
- Probability that the child is a son: 50% or 0.5
- Probability that the mother passes the $X^c$ allele: 50% or 0.5
- Combined probability: $0.5 \times 0.5 - 0.25$ or 25%

### Result

- The probability that a carrier woman and a normal man will have a color-blind son is 25%.

**Test Your Skills**

1. Describe the inheritance pattern of hemophilia A and hemophilia B. How does this pattern affect the prevalence of these disorders in males and females?
2. A geneticist is conducting a study on hemophilia C in a small population. Out of 200 individuals tested, 25 are found to have hemophilia C. What is the prevalence of hemophilia C in this population? Show your calculations.
3. A newborn baby boy is diagnosed with hemophilia B. His parents are both carriers of the disorder. Explain how this situation could occur genetically, and outline the probabilities involved in such a scenario.
4. John and Emily are planning to have a family. John's maternal grandfather had hemophilia A, but John's mother is not a carrier. What are the chances that their son will have hemophilia A?

## Understanding Gene Linkage through Large Sample Sizes

### Key Concepts of Gene Linkage

**Linkage and Gene Distance**

1. T. H. Morgan suggested that gene linkage is associated with the physical distance between genes on a chromosome. The closer the genes, the stronger the linkage, meaning they are less likely to be separated by crossing over during meiosis.

**Map Units**

2. Scientists use map units to measure the distances between linked genes, where one map unit corresponds to a 1% recombination rate between two genes. This unit helps map the exact locations and combinations of genes within a linkage group.

**Stability of Parental Combinations**

3. Morgan also noted that the parental combinations of linked genes typically remain unchanged during inheritance, unless crossing-over occurs.

### Importance of Large Sample Sizes in Linkage Studies

**Statistical Power**

- A large number of progeny in genetic studies provides more statistical power. This larger dataset allows researchers to observe a broader range of genetic combinations.

**Detection of Linkage**

- With more data, it becomes easier to detect deviations from expected independent assortment patterns that might indicate linkage. The larger the sample, the clearer the patterns of linkage become.

**Reliability of Results**

- Larger sample sizes ensure the reliability of linkage analysis, as they reduce the impact of random chance on the observed genetic combinations.

### Example of Gene Linkage in Humans

**Hypothetical Traits**

- Consider the traits of hair color and the ability to roll the tongue, with alleles brown hair (B) or blonde hair (b), and the ability to roll the tongue (T) or the inability to roll (t).

**Assumed Linkage**

- If these traits are linked (i.e., located close together on the same chromosome), and no crossing-over occurs, certain trait combinations (e.g., BT and bt) will be inherited together more frequently.

**Observations in Large Samples**

- In a large group, it may become evident that individuals with brown hair frequently can roll their tongues, while those with blonde hair often cannot. This pattern is more discernible in a larger sample size, indicating linkage between these traits.



## Techniques Employed for Embryonic Screening

Embryonic screening is a vital process used to detect genetic diseases or chromosomal abnormalities in embryos. Below are descriptions of two primary techniques used for this purpose:

### Amniocentesis

1. **Procedure Overview**
   - **Anesthetic Application**: A local anesthetic may be administered to the mother to alleviate discomfort.
   - **Fluid Extraction**: A needle is inserted through the mother's abdominal wall and uterus into the amniotic sac, guided by ultrasound to avoid contact with the fetus. Around 20ml of amniotic fluid, which contains fetal cells, is carefully withdrawn.
2. **Cell Processing**
   - **Cell Cultivation**: The fetal cells extracted with the amniotic fluid are cultured in a growth medium to increase their number.
   - **Chromosome Analysis**: Once sufficient cells are available, they are fixed and stained, then examined under a microscope to identify any chromosomal abnormalities, such as trisomy 21, which causes Down syndrome.

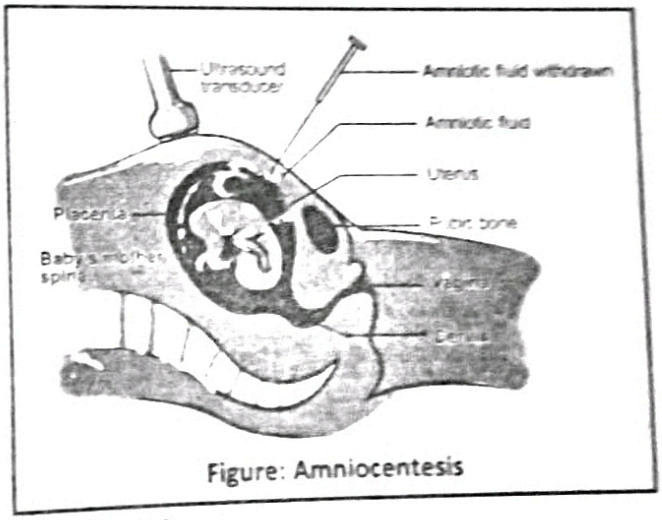


Figure: Amniocentesis

### Chorionic Villus Sampling (CVS)

1. **Procedure Overview**
   - **Tissue Collection**: Chorionic villus sampling involves collecting tissue from the chorionic villi, which are finger-like projections of placental tissue that share the fetus's genetic makeup.
   - **Sampling Methods**: The sample can be taken either transcervically, where a narrow tube is inserted through the cervix, or transabdominally, where a needle is passed through the abdominal wall into the placenta.

2. **Advantages and Safety**
   - **Rapid Results**: The rapidly dividing cells of the chorionic villi allow for immediate karyotyping, providing quick results to expecting parents.
   - **Safety and Efficacy**: When performed by a skilled practitioner with ultrasound guidance, CVS is a safe procedure with minimal complications, making it a reliable option for early diagnostic prenatal genetic testing.
3. **Decision-Making Support**:
   - **Informed Choices**: The results from CVS can help families make informed decisions regarding their pregnancy, based on potential genetic issues identified through the testing.

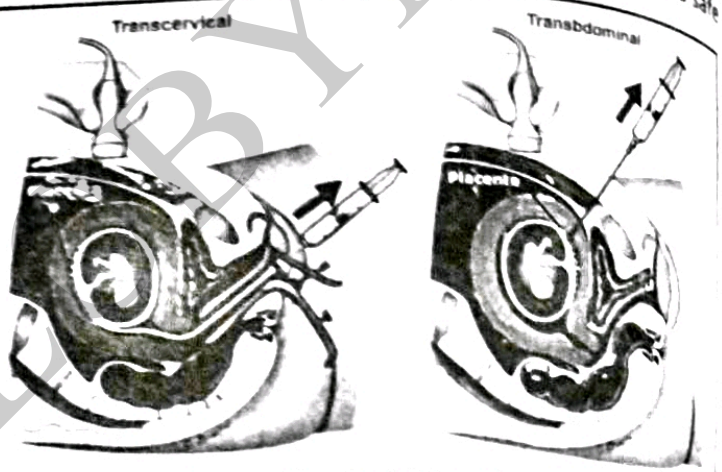



Figure: Chorionic Villus Sampling

- Both amniocentesis and chorionic villus sampling are crucial tools in prenatal genetic testing, offering families the ability to assess the genetic health of their future children and make informed decisions based on comprehensive genetic information.

**Test Your Skills**

1. Why chorionic villus sampling (CVS) provides rapid results compared to amniocentesis.
2. What biological factors contribute to this difference in timing?
3. A couple has a family history of cystic fibrosis and is concerned about their unborn child's risk of inheriting the condition.
4. Describe how chorionic villus sampling (CVS) could be used to test for cystic fibrosis in the fetus.
5. Describe the specific genetic mutation associated with cystic fibrosis and how it could be detected using CVS.
6. Describe the primary differences between amniocentesis and chorionic villus sampling (CVS) in terms of procedure and timing during pregnancy.