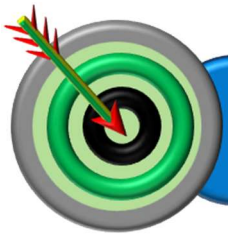
Chapter
04

Principles of Inheritance and Variation



OBJECTIVES

- INTRODUCTION
- MENDEL'S LAWS OF INHERITANCE
- INHERITANCE OF ONE GENE
- INHERITANCE OF TWO GENE
- POLYGENIC INHERITANCE
- PLEIOTROPY
- SEX DETERMINATION
- MUTATION
- GENETIC DISORDERS

INTRODUCTION

Reproduction is one of the most fundamental characteristics of all living organisms. It ensures the transfer of traits from parents to their offspring through reproductive cells like eggs and sperm. This transfer of traits results in offspring that resemble their parents, as seen when a mango seed grows into a mango tree or an elephant gives birth to a baby elephant. The transfer of characters from parents to offspring is known as inheritance. However, offspring are not identical to their parents in every aspect, leading to variations. Variation is the degree by which progeny differ from their parents. The branch of science which deals with the inheritance as well as the variation of characters from parents to offspring is Genetics.

Early agriculturists (8.000-10,000 B.C.) knew that one of the causes of variation was hidden in sexual reproduction. Because of it, they successfully bred domesticated varieties from wild plants and animals through selective crossing and artificial selection. Indian cow (e.g. Sahiwal of Punjab) is domesticated form of an ancestral wild cow. However, our ancestors had very little idea about the scientific basis of inheritance and variation.

4.1 MENDEL'S LAWS OF INHERITANCE

Gregor Johann Mendel was the first to scientifically explain the principles of inheritance and variation through his hybridization experiments. Experiments performed by Mendel on genetics and description of mechanisms of hereditary processes and formulation of principles are known as Mendelism. But it should be very much clear that he was not the first to conduct these experiments, rather he was the first to consider one to three characters at one time and this was perhaps the secret of his success.

Mendel conducted hybridisation experiments on garden pea (*Pisum sativum*) for seven years (1856 – 1863) and proposed laws of inheritance.

Before discussing, why did Mendel select pea plant for genetics experiment, we must know what is the difference between character and trait.

Character	Trait
Feature of the individual. e.g., Stem height, seed colour, seed shape etc.	Distinguishable feature of a character and its detectable variant. e.g., Tallness or dwarfness.

Selection of pea plant: The main reasons for adopting garden pea for experiments were as follows:

(1) Pea has many distinct alternative traits (clear contrasting traits).
(2) It produces a large number of seeds and completes its life cycle in one season.
(3) Flowers show self (bud) pollination, so are true breeding.
(4) It is easy to artificially cross-pollinate the pea flowers. The hybrids thus produced were fertile.

Mendel conducted artificial pollination/cross pollination experiments using several true-breeding pea lines.

A **true breeding line** is one that having undergone continuous self-pollination for several generations.

Mendel selected **14 true-breeding peas** plant varieties, as pairs which were similar except for one character with contrasting traits.. It means, Mendel selected 7 characters in pea plant for carrying out hybridisation experiments. These characters are listed in following table.

S.N.	Character	Contrasting traits	
		Dominant	Wrinkled
1.	Seed shape	Round	Wrinkled
2.	Seed colour	Yellow	Green
3.	Flower colour	Violet	White
4.	Pod shape	Full/Inflated	Constricted
5.	Pod colour	Green	Yellow
6.	Flower position	Axial	Terminal
7.	Stem height	Tall	Dwarf

Critical Thinking

1. Initially Mendel took 34 varieties of pea plants, then 22 but ultimately worked with only 7 pairs of varieties.
2. Term 'Pure line' was coined by Johannsen in 1900.

Reasons for Mendel's Success

(1) Mendel applied statistical method and mathematical logic for analysing his results.
(2) He kept accurate records of his experiments, giving all the details of number and type of individuals, which are a necessity in the genetic studies.
(3) Mendel experimented on a number of plants for the same trait and obtained hundreds of offspring. A large sampling size gave credibility to his results. Chances of error are little in large samples.
(4) He tried to formulate theoretical explanations for the observed results. These explanations were further tested by conducting experiments for successive generations of the test plants, that proved his results pointed to general rules of inheritance rather than being unsubstantiated ideas.

TOPIC CENTRIC EXERCISE -01

Q1. Some of the dominant traits studied by Mendel were
(a) Round seed shape, green seed colour, and axial flower position.
(b) Terminal flower position, green pod colour, and inflated pod shape.
(c) Violet flower colour, green pod colour, and round seed shape.
(d) Wrinkled seed shape, yellow pod colour, and axial flower position

Q2. Mendel selected how many true breeding pea plant?
(a) 12 (b) 13
(c) 14 (d) 15

Q3. Correct reason for Mendel's success was
(a) He repeated each experiment several times
(b) Traits chosen by him had genes far apart so that linkage was absent
(c) He kept record of all experiments
(d) He used statistical techniques

Q4. Gregor Mendel conducted hybridisation experiments for
(a) Seven years (1865-1872) (b) Seven years (1856-1863)
(c) Seven years (1853-1860) (d) Fourteen years (1853-1867)

Q5. Which one is not a dominant trait amongst seven pea traits chosen by Mendel?
(a) Flower Colour-Violet (b) Pod Colour-Yellow
(c) Shape of Seed-Round (d) Flower-Axial

4.2. INHERITANCE OF ONE GENE

When the inheritance of one character is considered at a time in a cross this is called monohybrid cross. Study of inheritance of single pair of contrasting traits of a character at a time is called **one gene inheritance**. Mendel crossed tall and dwarf pea plants to study the inheritance of one gene. He collected the seeds produced as a result of this cross and grew them to generate plants of the first hybrid generation. This generation is also called the **filial$_1$, (offspring) progeny** or the $\boldsymbol{F_1}$.

Mendel found that all F_1 progeny plants were tall, like one of its parents; none were dwarf. He made one similar cross with other pairs of contrasting traits and in every case the result was the same. In each, F_1 plants were identical to one of the parents.

S. No.	Characters	Parents		F_1 Plants
1.	Stem height	Tall	Dwarf	Tall
2.	Flower colour	Violet	White	Violet
3.	Flower position	Axial	Terminal	Axial
4.	Pod shape	Inflated	Constricted	Inflated
5.	Pod colour	Green	Yellow	Green
6.	Seed shape	Round	Wrinkled	Round
7.	Seed colour	Yellow	Green	Yellow

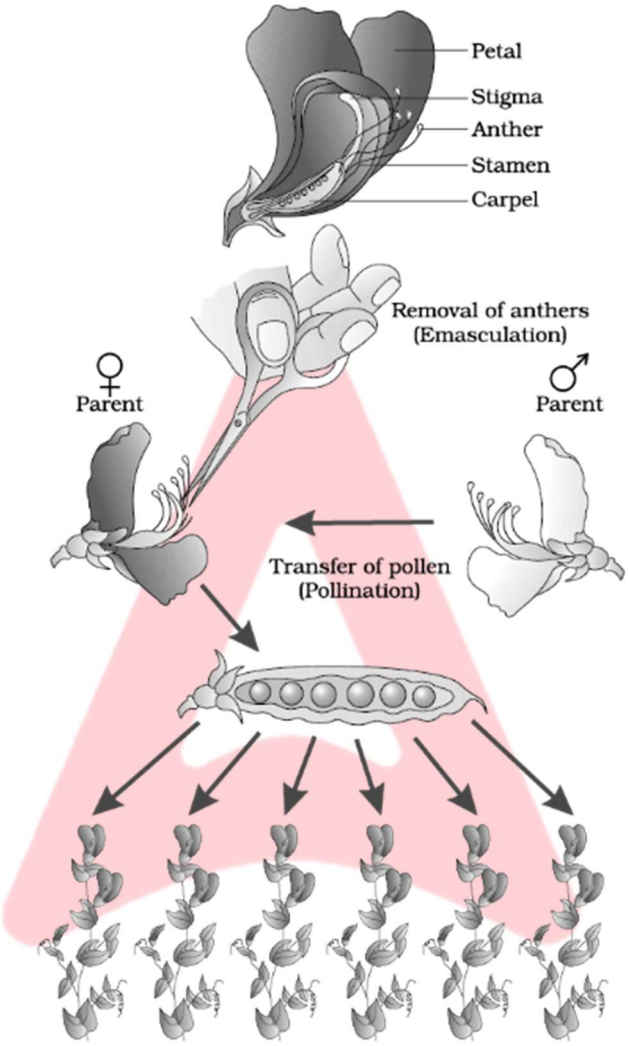


Fig.: Steps in making a cross in pea

When Mendel self-pollinated the tall F_1 plants, both tall and dwarf plants were obtained in F_2 generation. Offspring derived from selfing of the F_1 are termed as **second filial** or $\boldsymbol{F_2}$ **generation**. The proportion of plants that were tall were $\left(\frac{3}{4}\right)^{th}$ of the F_2 population while $\left(\frac{1}{4}\right)^{th}$ of the F_2 population were dwarf. We must note here that dwarfness which disappeared in F_1 generation, reappeared in F_2. The tall and dwarf traits in F_2 generation were identical to their parental type and **did not show any blending**, i.e., all the offsprings were either tall or dwarf, **none were of in-between height**.

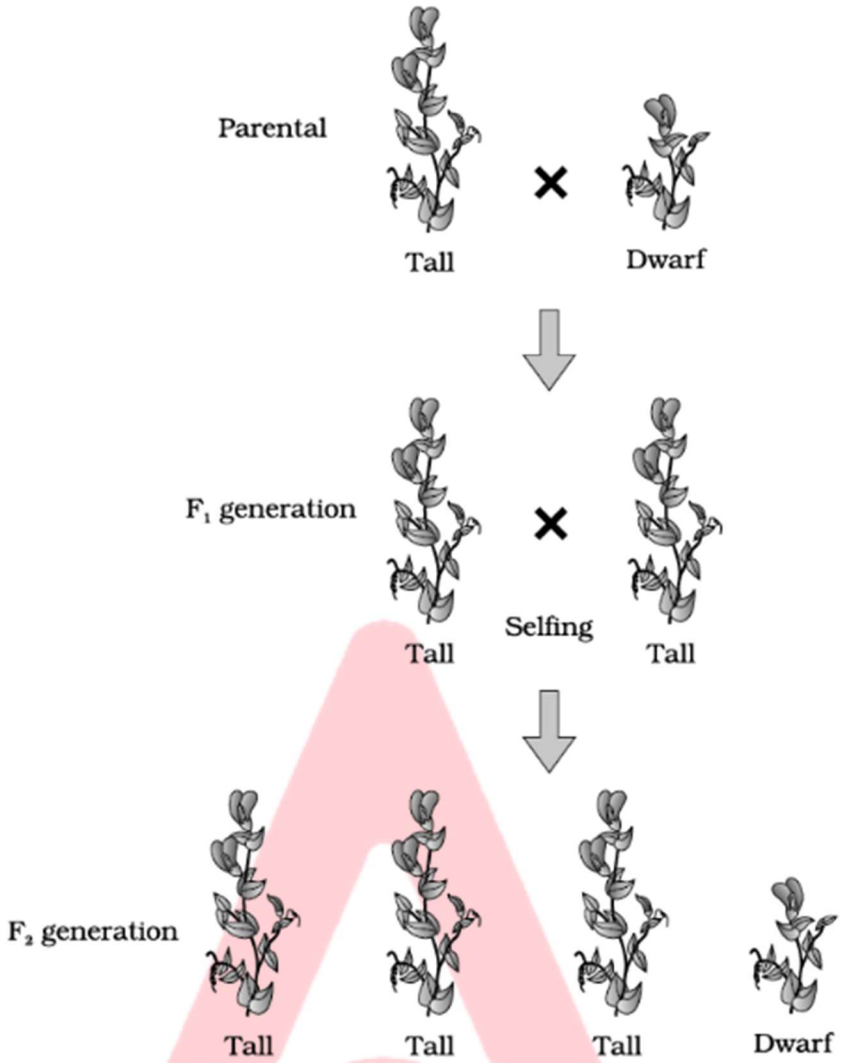


Fig.: Diagrammatic representation of monohybrid cross

He made similar crosses with other pairs of contrasting traits and observed the F_2 generation in which both the traits were expressed in the proportion of 3: 1

- **Concept of 'Factors'**

Based on these observations, Mendel proposed that something was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as **'factors'**. We now call these factors as **"genes"**. Therefore, a gene is defined as the functional unit of inheritance. They contain the information that is required to express a particular trait in an organism. Chemically gene is a segment of DNA that has a particular function, the common being synthesis of polypeptide.

Genes which code for a pair of contrasting traits are known as **alleles** i.e., they are slightly different forms of the same gene. Therefore, term gene can be used for any factor but term allele is used with reference to another allele. We use alphabetical symbols for each gene, the capital letter is used for the trait expressed at the F_1 stage and small alphabet for the other trait. For example, if **T** is used for the 'tall' trait and **t** for 'dwarf' then **T** and **t** are alleles of each other. Therefore, in plants (Diploid) the pair of alleles for height would be **TT, Tt** or **tt**

Critical Thinking

1. Term 'gene' was given by Johannsen while term 'allele' by Bateson.
2. Alleles are the abbreviated form of the term "allelomorphs".

- **Homozygous and Heterozygous**

  Homozygous- It is an individual which contains identical alleles of a gene or factor of a character on its homologous chromosome. E.g., TT and tt.

  Heterozygous- It is an individual which contains the two contrasting factor of a character or two different alleles of a gene on its homologous chromosomes. E.g., Tt

- **Genotype and Phenotype**

  Genotype is representation of genetic complement of an individual with respect to one or more characters. e.g, TT, Tt, tt**.**

  Phenotype is an individual which contains the two contrasting factor of a character or two different alleles of a gene on its homologous chromosomes. E.g., Tt.

- **Dominant and Recessive**

  Based on the results obtained in F_1 generation, Mendel was able to propose that when two dissimilar factors are present in a single individual, only one is able to express and the other is not. The one that expresses itself is called **dominant** factor while which fails to express is termed as **recessive** factor. In other words we can say that a dominant allele influences the appearance of the phenotype even in the presence of an alternative allele while recessive allele influences the appearance of the phenotype only in the presence of another identical allele.

- **Concept of Segregation**

  From the above observation that the recessive parental trait (dwarfness, tt) is expressed without any blending in the F_2 generation, we can infer that, when the tall and dwarf plant produces gametes by the process of meiosis, the alleles of the parental pair separate (segregate) from each other and only one allele is transmitted to a gamete. It means meiosis reduces the number of chromosomes to one half where a gamete carries only one chromosome of each type and hence only one factor of a character.

  The segregation of alleles is a random process and so there is a 50 percent chance of a gamete containing either allele. In this way the gametes of the tall **TT** plants have the allele **T** and the gametes of the dwarf **tt** plants have the allele **t**. During fertilization of the two alleles, **T** from one parent through the pollen (**n**), and **t** from the female parent through the egg (**n**) are united to produce zygotes (2n) that have one **T** allele and one **t** allele i.e. hybrid or heterozygous **Tt** plant (2n).

Critical Thinking

Type of gametes produced by a diploid individual can be calculated by using formula, 2^n. Here 'n' represents the number of heterozygotes/hybrid.

- **Punnett Square**

  The production of gametes(n) by the parents (2n), the formation of the zygotes (2n), the F_1 and F_2 plants can be understood from a diagram called **Punnett square**. Punnett square was developed by a British geneticist, Reginald C. Punnett. It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross. The possible gametes are written on two sides, male in horizontal row and female in vertical column. All possible combinations are represented in boxes below in the squares, which generates a square output form.

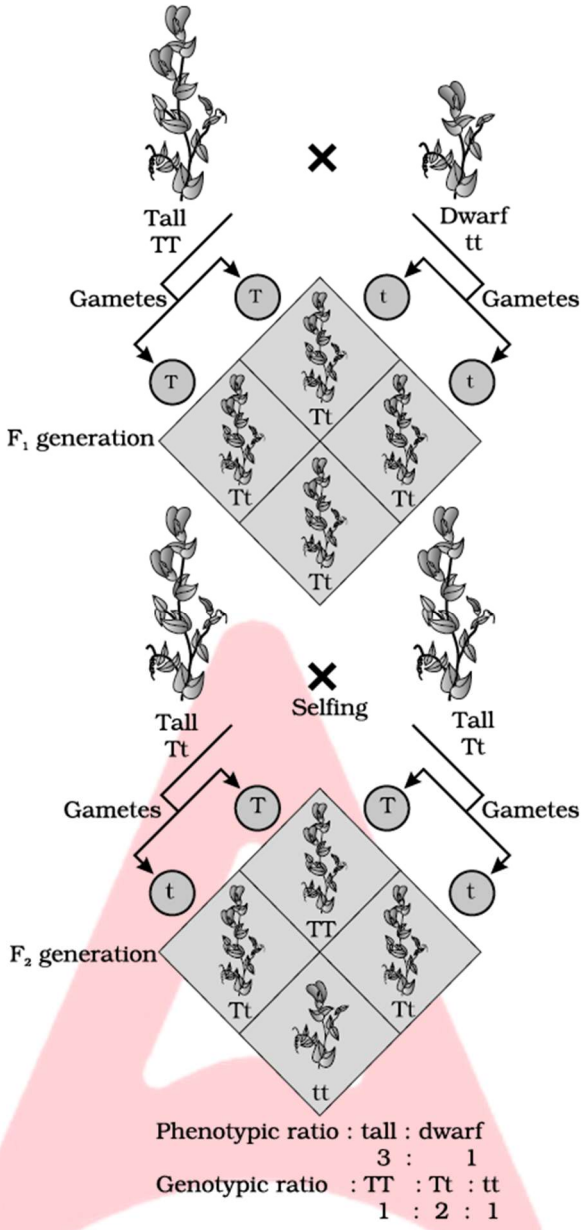


Fig.: A Punnett square used to understand a typical monohybrid cross conducted by Mendel between true-breeding tall plants and true-breeding dwarf plants

The Punnett square, given in above figure, shows the parental tall **TT** (male) and dwarf **tt** (female) plants, the gametes produced by them and the $\boldsymbol{F_1}$ **(Tt)** progeny. The $\boldsymbol{F_1}$ plant of the genotype **Tt** when self-pollinated, produces gametes of the genotype **T** and **t** in equal proportion. When fertilization takes place, the pollen grains of genotype T have a 50% chance to pollinate eggs of the genotype **T**, as well as of genotype **t**. Also pollen grains of genotype t have a 50% chance to pollinate eggs of genotype **T**, as well as of genotype **t**. As a result of random fertilisation, the resultant zygotes can be of the genotypes **TT, Tt** or **tt**. From the Punnett square it is easily seen that $1/4^{th}$ of the random fertilisations leads to **TT**, ½ lead to **Tt** and $1/4^{th}$ to **tt**. Due to dominance of one trait over the other that all the F_1 are tall (though the genotype is **Tt**) and in the F_2, $3/4^{th}$ of the plants are tall (though genotypically ½ are **Tt** and $1/4^{th}$ are **TT**). This leads to a phenotypic ratio of $3/4^{th}$ tall : (¼ **TT** + ½ **Tt**) and $1/4^{th}$ tt, i.e., a 3:1 ratio, but a genotypic ratio of 1:2:1. The ¼ : ½ : ¼ ratio of **TT : Tt : tt** is mathematically condensable to the form of the binomial expression $(ax + by)^2$, that has the gametes bearing genes **T** or **t** in equal frequency of ½.

The expression is expanded as given below:

$$\left(\frac{1}{2}T+\frac{1}{2}t\right)^2=\left(\frac{1}{2}T+\frac{1}{2}t\right)\times\left(\frac{1}{2}T+\frac{1}{2}t\right)=\frac{1}{4}TT+\frac{1}{2}Tt+\frac{1}{4}tt$$

Mendel self-pollinated the F_2 plants and found that dwarf F_2 plants continued to generate dwarf plants in F_3 and F_4 generations. He concluded that the genotype of the dwarfs was homozygous i.e., **tt**.

➢ **Test Cross**

When F_1 progeny is crossed with recessive parent then it is called test cross. The total generations obtained from this cross, 50% having dominant character and 50% having recessive character.

Monohybrid Test Cross: The progeny obtained from the monohybrid test cross are in equal proportion , means 50% is dominant phenotypes and 50% is recessive phenotypes.

Monohybrid test cross ratio = 1:1

Example: It we want to determine the genotype of a violet-flowered pea plant (test organism), then it is crossed with the recessive parent (white-flowered pea plant) instead of self-crossing.

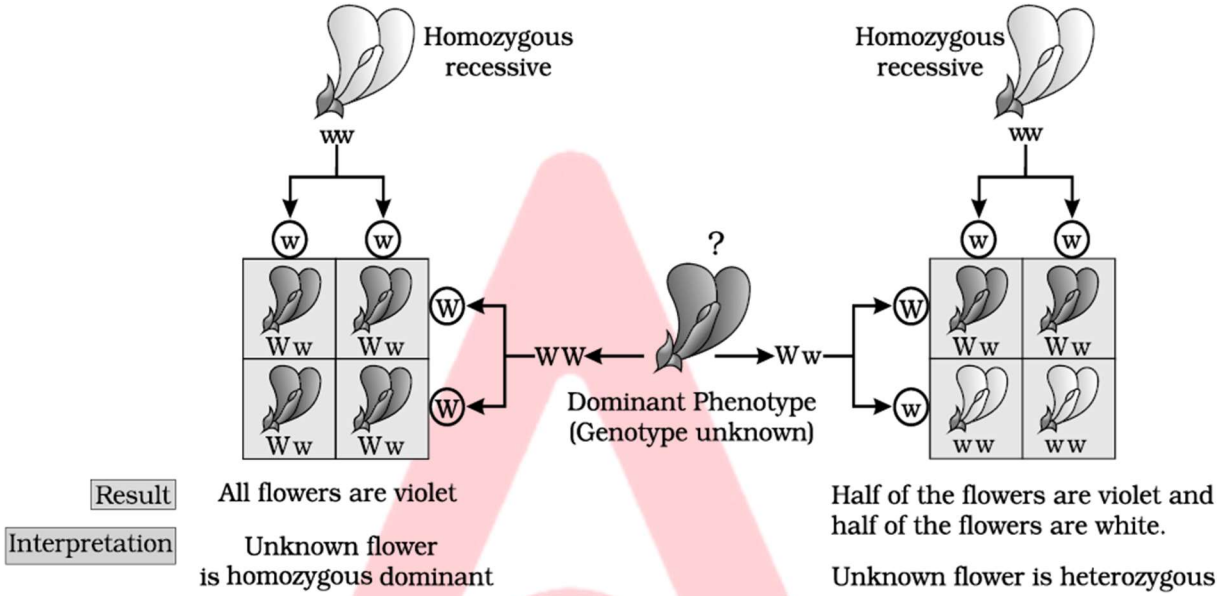


Fig.: Diagrammatic representation of a test cross

Clue Finder

- In test cross, phenotypes and genotypes ratio are same.
- The Punnett square can be effectively used to understand the independent segregation of the two pairs of genes during meiosis and the production of eggs and pollen in the plants.

On the basis of his observations on monohybrid cross, Mendel proposed two general rules. Today these rules are called the principles or Laws of Inheritance:

1. The first law or Law of Dominance
2. The second law or Law of Segregation

4.2.1 Law of Dominance:

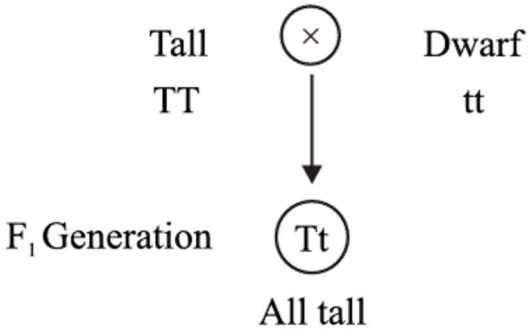
Mendel experimented with garden pea for seven characters. In each case he found that:

(i) Every character is controlled by discrete units called factors.

(ii) The factors occur in pairs.

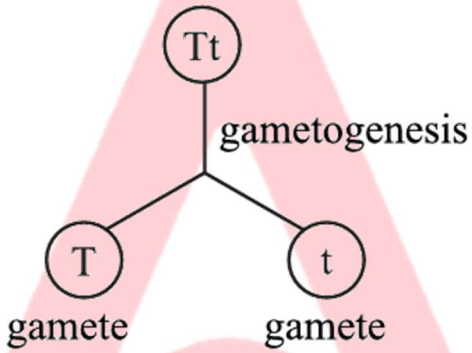
(iii) In a dissimilar pair of factors (e.g. Tt), only one is able to express its effect that is called as dominant factor. The other factor which does not show its effect is known as recessive factor.

The law of dominance is used to explain the expression of only one of the parental traits in a monohybrid cross in the F_1 and the expression of both in the F_2. It also explains the proportion of 3: 1 obtained in F_2 generation. This law is not universally applicable.



4.2.2 Law of Segregation:

This law is based on the fact that the two factors of a character present in an individual do not get mixed up (blending) and both the traits are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage. During gamete or spore formation, factors of a pair separate or seg-regate from each other, so that a gamete carries only one factor of a character. This ensures the purity of gametes. Of course, a homozygous parent produces all gametes that are similar while a heterozygous will produce two type of gametes each having one factor with equal proportion. This law is universally applicable.



Exceptions to Mendelian Principles: Exception to mendelian principles are- Incomplete Dominance and Codominance

4.2.3 Incomplete Dominance:

After Mendelism, a few cases were observed where F_1 phenotype is intermediate between dominant and recessive phenotype, it means F_1 did not resemble either of the two parents and was in between the two.

A good example of incomplete dominance is that of flower colour in Snapdragon (dog flower or *Antirrhinum majus*). True-breeding red-flowered plant **(RR)** was crossed with true breeding white-flowered plant (rr). F_1 offspring **(Rr)** had pink flowers. Here one allele is incompletely dominant over other so that intermediate phenotype is produced by F_1 hybrid with respect to the parents. If the F_1 is selfed, the plants of F_2 generation are of three types red **(RR)**, pink **(Rr)** and white flowered **(rr)** in the ratio of **1: 2: 1**. In heterozygous condition **(Rr)**, phenotypic effect of one allele is more pronounced than that of other and then mixing of both colours (red & white) results in the development of pink colour.

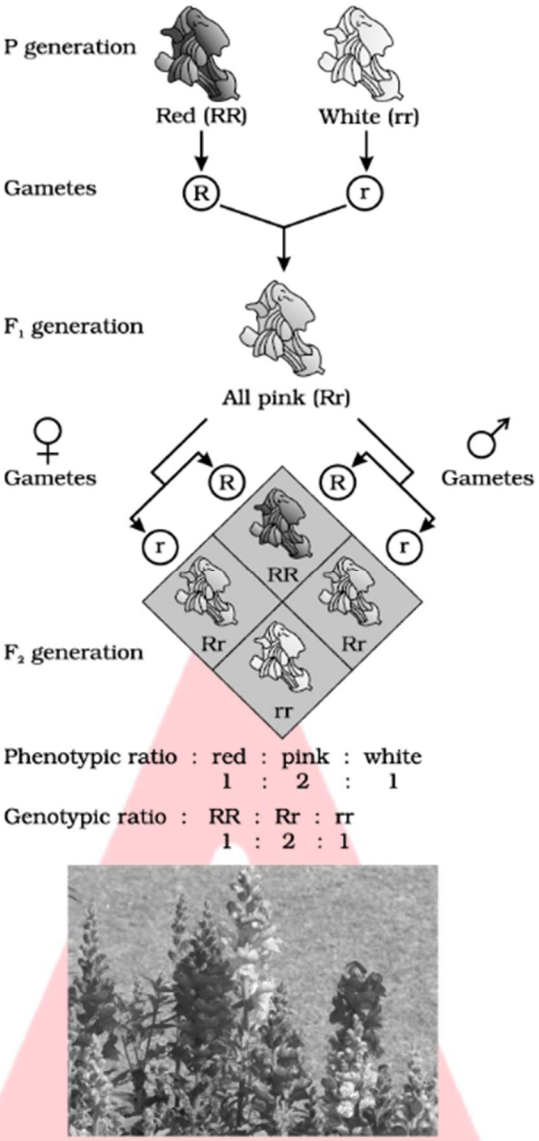


Fig.: Results of monohybrid cross in the plant Snapdragon

The Mendelian concept of a gene controlling a single character has also expanded to take into account genes which affect several characters simultaneously (pleiotropy). It means in pleiotropy, **a single gene product may produce more than one effect** or control several phenotypes depending on its position. The basis of pleiotropy is the interrelationship between the metabolic pathways that may contribute towards different phenotypes. Examples:

(a) In phenylketonuria, mutation of a gene that codes for the enzyme phenylalanine hydroxylase, results in a phenotypic expression characterised by mental retardation and a reduction in hair and skin pigmentation.

(b) The gene controlling starch synthesis in garden pea. It has two alleles, **B** and **b**. Starch synthesis in **BB** homozygotes is efficient and therefore large starch grains are produced. In **bb** homozygotes, starch synthesis is less efficient, so that it produces small-sized starch grains. After maturation of seeds, **BB** seeds are round and **bb** seeds are wrinkled. Heterozygotes **Bb** form round seeds but the starch grains are of intermediate size. Now it is clear that if starch synthesis is considered, **Bb** seeds show incomplete dominance but if seed shape is considered, **B** allele is dominant and **b** is recessive.

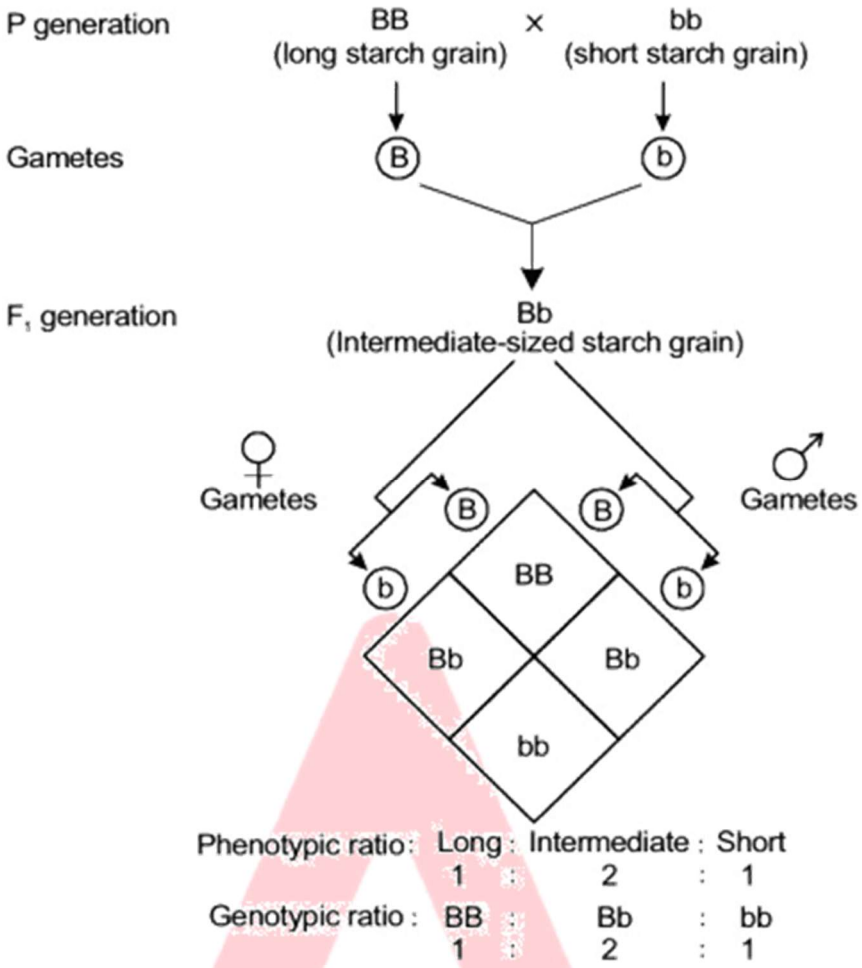


Fig.: Results of monohybrid cross in garden pea for size of starch grain

Clue Finder

Dominance is not an autonomous feature of a gene or the product. It depends upon the gene product and particular phenotype we choose to examine when a gene produces more than one phenotype.

Explanation of the Concept of Dominance:

Each gene carries the information required to express a specific trait. In diploid organisms, there are two copies of each gene, known as alleles. These alleles can be either identical or different. If they differ, it could be due to a mutation—a sudden change in the genetic makeup that alters the information carried by the allele. For example, a normal allele might produce an enzyme necessary to convert a substrate "S.". Theoretically, the modified or mutated allele could be responsible for production of

(i) The normal/less efficient enzyme, or
(ii) A non-functional enzyme, or
(iii) No enzyme at all

In case (i), the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait as in case of silent mutation. But, if the allele produces a non-functional enzyme or no enzyme [case (ii) & (iii)], the phenotype may be affected. The **unmodified (functioning) allele**, which represents the original phenotype is the **dominant allele/wild type** and the **modified allele** is **generally** the **recessive allele/mutant type**. Hence, the recessive trait is due to non-functional enzyme or because no enzyme is produced.

4.2.4 Co-dominance:

Besides incomplete dominance, certain alleles show co-dominance. Here in F_1 hybrid, both alleles express themselves equally and there is no mixing of the effect of the both alleles, therefore **hybrid progeny (F_1) resembles both parents**. The alleles which do not show dominance-recessive relationship and are able to express themselves independently when present together are called co-dominant alleles.

ABO blood group is also a good example of co-dominance. ABO blood groups are controlled by the gene *I*. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene (I) has three alleles ***I^A, I^B, i***. The alleles $\mathbf{I^A}$ and $\mathbf{I^B}$ produce a slightly different form of the sugar while allele i does not produce any sugar or antigen. $\mathbf{I^A}$ and $\mathbf{I^B}$ are completely dominant over **i**, in other words when $\mathbf{I^A}$ and **i** are present only $\mathbf{I^A}$ expresses as **i** does not produce any sugar, and when $\mathbf{I^A}$ and **i** are present, only $\mathbf{I^A}$ expresses. When both $\mathbf{I^A}$ and $\mathbf{I^B}$ are present in a person, both enzymes or sugars thus both antigens **A** and **B** are produced. This is because of co-dominance. These antigens determines the type of blood group. Blood group **A** have antigen **A**, group **B** have antigen **B, AB** have both antigens while blood group **O** do not carry any antigen. Thus, six genotypes and four phenotypes are possible.

Table: Genetic Basis of Blood Groups in Human Population

Allen from Parent 1	Allen from Parent 2	Genotype of offspring	Antigen	Blood types of offspring
I^A	I^A	I^AI^A	A	A
I^A	I^B	I^AI^B	A, B	AB
I^A	i	$I^A\,i$	A	A
I^B	I^A	I^AI^B	A, B	AB
I^B	I^B	I^BI^B	B	B
I^B	i	$I^B\,i$	B	B
i	i	ii	Neither	O

Other example of co-dominance is - Carrier of Sickle cell anaemia ($Hb^A\ Hb^S$)

- **Multiple allelism:** Mendel proposed that each gene has two contrasting forms i.e., alleles. But there are some genes which are having more than two alternative forms (allele). Presence of more than two alleles for a gene is known as multiple allelism.

A good example is different types of red blood cells that determine ABO blood grouping in human beings. ABO blood groups are controlled by the gene **I**. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene **(I)** has three **alleles $\mathbf{I^A}$, $\mathbf{I^B}$** and $\mathbf{i/I^O}$. Despite the presence of three alleles of the same gene in a population, an individual (2n) can have only two alleles. Therefore, multiple alleles can be detected **only in a population.** Since there are three different alleles, therefore six different genotypes are possible for this character (I^AI^A, I^AI^O; I^BI^B, I^BI^O; I^AI^B; I^OI^O or ii).

TOPIC CENTRIC EXERCISE -02

Q1. According to Sutton and Boveri, what leads to the segregation of a pair of factors carried by chromosomes?
(a) Genetic modification
(b) Natural selection
(c) Pairing and separation of chromosomes
(d) Environmental factors

Q2. Punnett square is used to know
(a) Outcome of a cross (b) Probable result of a cross
(c) Type of gametes (d) Result of meiosis

Q3. Which of the following statements about co-dominance is true?
(a) It results in a mix of traits from both parents.
(b) It occurs when one allele is completely dominant over the other.
(c) It occurs when two alleles express their own traits simultaneously

(d) It leads to an incomplete expression of traits.

Q4. In a monohybrid cross between two heterozygous individuals, percentage of pure homozygous individuals obtained in F_1, generation is

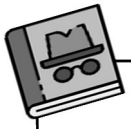
(a) 25% (b) 50%

(c) 75% (d) 100%

Q5. When both alleles express their effect on being present together, the phenomenon is called:

(a) Dominance (b) Incomplete dominance

(c) Pseudodominance (d) Co-dominance



Clue Finder

If n is the number of alleles of a gene then number of different possible genotype = $\frac{n(n+1)}{2}$

4.3 INHERITANCE OF TWO GENE

It is a cross between two organisms of a species that are different in two pairs of contrasting characters.

Mendel wanted to observe the effect of one pair of heterozygous on other pair. So, he selected traits for dihybrid cross for his experiment as follows:

(i) Colour of seeds → Yellow (Y) and Green (y)

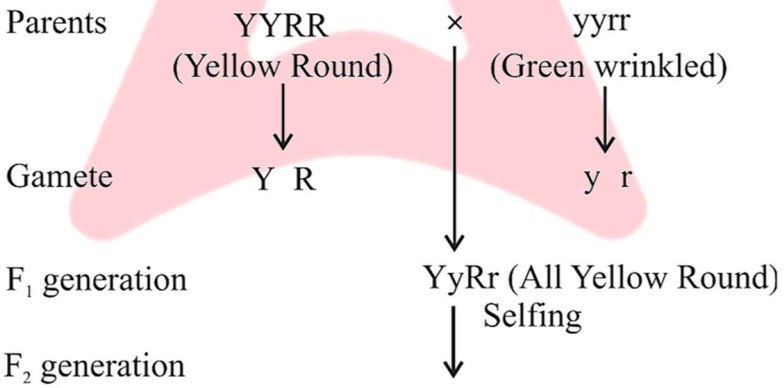
(ii) Seed shape → Round (R) and Wrinkled (r)

Yellow and round characters are dominant and green and wrinkled are recessive characters.

All the plants of the F_1 generation were yellow and round seeds (YyRr).

The factors of both characters will have independently segregated to each other during gamete formation. Thus total four types of gametes (YR), (yR), (Yr), (yr) form in F_1 generation.

On selfing of F_1 the resultant F_2 generation show four types of plants, i.e., Yellow Round, Yellow wrinkled, Green Round, Green wrinkled



♀ ♂	YR	yR	Yr	Yr
YR	YYRR (Yellow Round)	YyRR (Yellow Round)	YYRr (Yellow Round)	YyRr (Yellow Round)
yR	YyRR (Yellow Round)	yyRR (Green Round)	YyRr (Yellow Round)	YyRr (Green Round)
Yr	YYRr (Yellow Round)	YyRr (Yellow Round)	YYrr (Yellow wrinkled)	Yyrr (Yellow wrinkled)
yr	YyRr (Yellow Round)	yyRr (Green Round)	Yyrr (Yellow wrinkled)	yyrr (Green wrinkled)

Phenotypic ratio	9	:	3	:	3	:	1									
	Yellow Round		Yellow wrinkled		Green Round		Green wrinkled									
Genotypic ratio	1	:	2	:	2	:	4	:	1	:	2	:	1	:	2	: 1
	YYRR		YYRr		YyRR		YyRr		YYrr		Yyrr		yyRR		yyRr	yyrr

4.3.1 Law of Independent Assortment

Based upon the results obtained in dihybrid crosses, Mendel proposed a second set of generalisations that we call **Mendel's Law of Independent Assortment**. The law states that "when two pairs of traits are combined in a hybrid, segregation of one pair of traits is independent to the other pair of traits".

The Punnett square can be effectively used to understand the independent segregation of the two pairs of factors during meiosis and the production of eggs (haploid) and pollen (haploid) in the F_1, (RrYy) plant (diploid).

If we consider the segregation of one pair of factors R and r, 50% of the gametes have the factor R and the other 50% have r. Along with R or r in the gametes, it should also have the factor Y or y. Here, it is important thing to remember that segregation of 50% R and 50% r is independent from the segregation of 50% Y and 50% y. Therefore, 50% of the r bearing gametes has Y and the other 50% has y. Similarly, 50% of the R bearing gametes has Y and the other 50% has y. Thus, there are 4 genotypes of gametes (four types of pollen and four types of eggs). The four types are RY, Ry, rY and ry each with a frequency of 25% or 1/4th of the total gametes produced. If we write down the four types of pollens and eggs on the two sides of a Punnett square it is very easy to derive the composition of zygotes that give rise to the F_2 plants.

4.3.2 Chromosomal Theory of Inheritance

Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900.

➢ **Mendel Results Remain Hidden Due To**

- Communication was not easy (as it is now) in those days and his work could not be widely publicized.
- His concept of **genes** (or **factors**, in Mendel's words) as stable and discrete units that controlled the expression of traits and, of the pair of alleles which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.
- Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.
- Though Mendel's work suggested that factors (genes) were discrete units, he could not provide any physical proof for the existence of factors or say what they were made of.

Critical Thinking

Reasons for Mendel's Success-

Mendel studied the **inheritance** of one or two characters at a time unlike his predecessors who had considered many characters at a time.

The advancements in microscopy led to the discovery of structures in the nucleus that appeared to double and divide just before each cell division. These were called **chromosomes** (*colored bodies*, as they were visualised by staining).

It was proposed by **Sutton and Boveri** independently in **1902** and expanded by **Morgan, Sturtevant and Bridges**. Walter Sutton and Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws. The two alleles of a gene pair are located on homologous sites on homologous chromosomes.

You have studied the behaviour of chromosomes during mitosis (equational division) and during meiosis (reduction division). The important things to remember are that chromosomes as well as genes occur in pairs. The two alleles of a gene pair are located on homologous sites on homologous chromosomes.

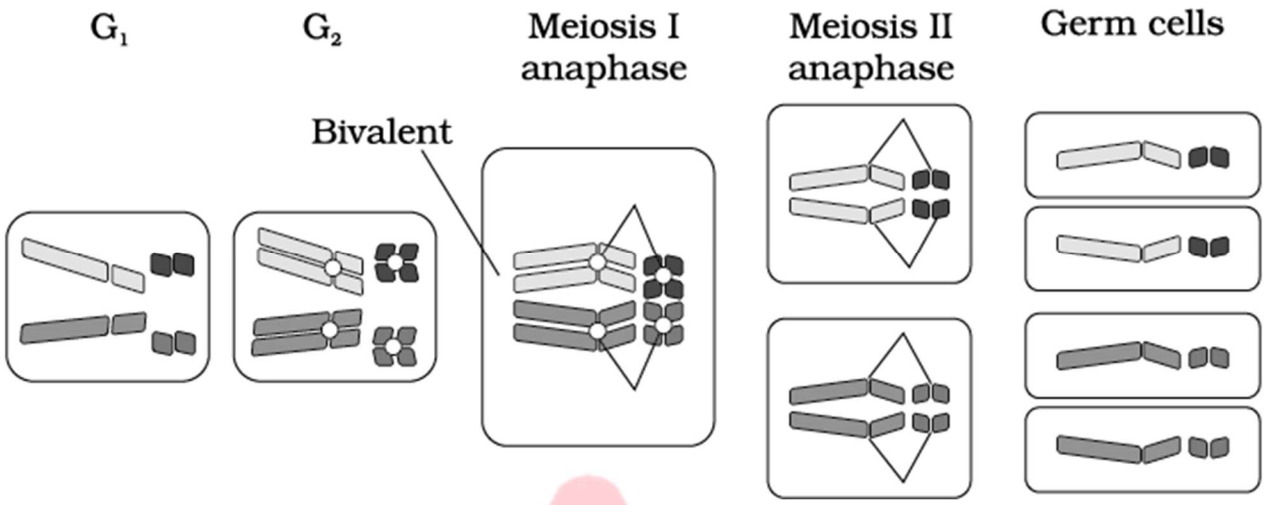


Fig.: Meiosis and germ cell formation in a cell with four chromosomes

During Anaphase of meiosis I, the two chromosome pairs can align at the metaphase plate independently of each other.

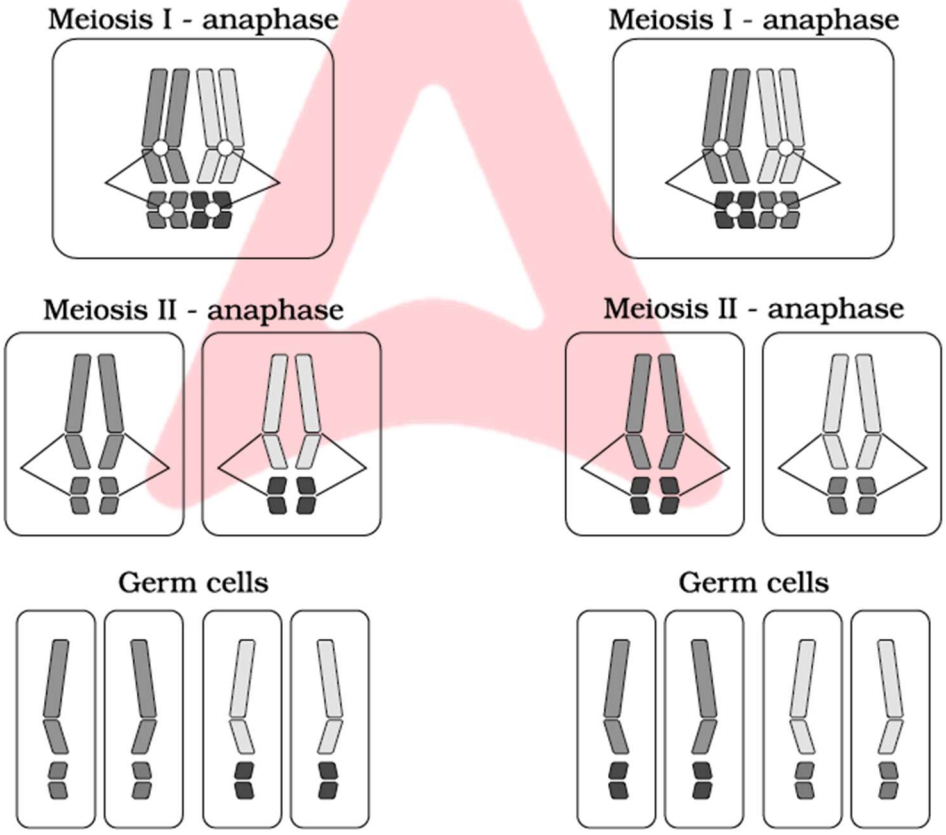


Fig.: Independent assortment of chromosomes

Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the **chromosomal theory of inheritance**.

Main features of chromosome theory of inheritance are as follows:

(i) Genes are present on chromosomes

(ii) Like the hereditary traits the chromosomes retain their number, structure and individuality throughout the life of an organism and from generation to generation. The two neither get lost nor mixed up. They behave as units.

(iii) Both chromosomes as well as genes occur in pairs in the somatic or diploid cells. The two alleles of a gene pair are located on homologous sites on homologous chromosomes. Both chromosomes as well as genes segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete.

(iv) A gamete contains only one chromosome of a type and only one of the two alleles of a trait.

(v) The paired condition of both chromosomes as well as Mendelian factors is restored during fertilization.

Thus, homologous chromosomes synapse during meiosis and then separate or segregate independently into different cells which establishes to quantitative basis for segregation and independent assortment of hereditary factors.

➢ **Experimental Verification of the Chromosomal Theory of Inheritance**

It was given by Thomas Hunt Morgan and his colleagues.

This led to discovering the basis for the variation that sexual reproduction produced.

Morgan worked with the tiny fruit flies, Drosophila *melanogaster* which were found very suitable for such studies.

➢ **Advantages of Selecting Drosophila melanogaster**

- They could be grown on simple synthetic medium in the laboratory.
- They complete their life cycle in about two weeks.
- A single mating could produce a large number of progeny flies.
- There was a clear differentiation of the sexes the male and female flies are easily distinguishable.
- It has many types of hereditary variations that can be seen with low power microscopes.

4.3.3 Linkage and Recombination

Linkage is defined as the co-inheritance of two or more non- allelic genes due to their being located more or less closely on the same chromosome.

Sex linkage was first discovered by **Morgan** in Drosophila & coined the term linkage. He proposed the theory of linkage.

Morgan carried out several dihybrid crosses in Drosophila to study genes that were sex-linked. The crosses were similar to the dihybrid crosses carried out by Mendel in peas. Morgan hybridised yellow-bodied, white-eyed females to brown-bodied, red-eyed males and intercrossed their F_1 progeny.

He observed that the two genes did not segregate independently of each other and the F_2 ratio deviated very significantly from the 9:3:3:1 ratio (expected when the two genes are independent). Morgan and his group knew that the genes were located on the X chromosome and saw quickly that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations were much higher than the non-parental type.

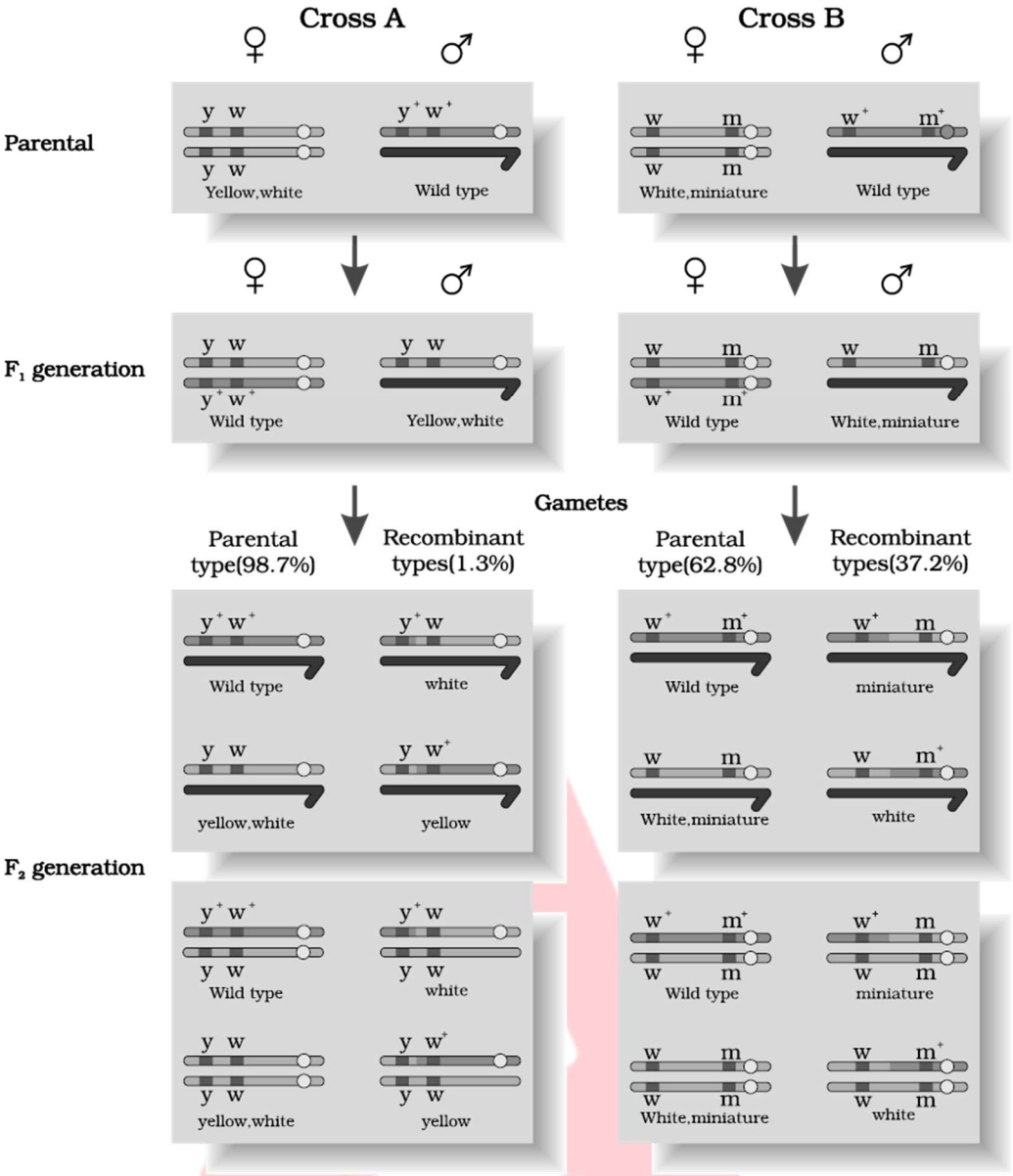
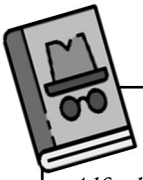


Fig.: Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript

Morgan attributed this due to the physical association or linkage of the two genes and coined the term linkage to describe this physical association of genes on a chromosome and the term recombination to describe the generation of non- parental gene combinations. Morgan and his group also found that even when genes were grouped on the same chromosome, some genes were very tightly linked (showed very low recombination) while others were loosely linked (showed higher recombination).



Clue Finder

Alfred Sturtevant (student of Morgan) used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and 'mapped' their position on the chromosome.

Linkage Maps or Chromosome Maps

The graphical representation of sequence and relative distance between genes in a chromosome is called linkage map.

The first linkage map was developed by **Sturtevant (1911)** for *Drosophila*.

The frequency of crossing over between two genes is directly proportional to the distance between the two. **Recombination frequency** or **Cross-over value (COV)** is measured by test cross. 1% crossing over between two linked genes is known as 1 map unit or 1 centimorgan.

$$\text{Recombination frequency} = \frac{\text{Number of recombinants}}{\text{Total number of off springs}} \times 100$$

Today genetic maps are extensively used as a starting point in the sequencing of whole genomes as was done in the case of the Human Genome Sequencing Project.

TOPIC CENTRIC EXERCISE -03

Q1. The ratio of 9:3:3: 1 is due to:
(a) Independent assortment of genes
(b) Segregation of characters
(c) Crossing over of chromosomes
(d) Homologous pairing between chromosomes

Q2. What is ratio of homozygous plants for both dominant characters in F_2 of a dihybrid cross?
(a) 1/16 (b) 3/16
(c) 4/16 (d) 9/16

Q3. The percentage of ab gamete produced by AaBb parent will be
(a) 25% (b) 50%
(c) 75% (d) 12.5%

Q4. Genetic map is one that
(a) Shows stages of cell division
(b) Establishes various stages seen in gene evolution
(c) Establishes sites of genes on a chromosome
(d) Shows distribution of various chromosomes in a cell

Q5. According to the law of independent assortment in a dihybrid cross
(a) There are four genotype in F_2
(b) F_2 contains 16 phenotypes
(c) There is a single individual which is homozygous recessive for both the characters
(d) It is not possible to forecast the different phenotypes

4.4 POLYGENIC INHERITANCE

Mendel's studies mainly described those traits that have distinct alternate forms such as flower colour which are either purple or white. But if you look around you will find that there are many traits which are not so distinct in their occurrence and are spread across a gradient. For example, in humans we do not just have tall or short people as two distinct alternatives but a whole range of possible heights. Such traits are generally controlled by two or more genes and are thus called as polygenic traits. The inheritance of polygenic traits is called polygenic or quantitative inheritance.

In quantitative inheritance, the dominant alleles have cumulative effect, with each dominant allele expressing a part of functional polypeptide and full trait is shown when all the dominant alleles are present. Genes involved in quantitative inheritance are called polygenes.

Example:

Human skin colour: The inheritance of colour of skin in human studied by Devenport. Seven types of phenotype of skin colour are found in human.

4.5 PLEIOTROPY

A single gene can exhibit multiple phenotypic expression. Such a gene is called a pleiotropic gene. The underlying mechanism of pleiotropy in most cases is the effect of a gene on metabolic pathways which contribute towards different phenotypes.

An example of this is the disease phenylketonuria, which occurs in humans. The disease is caused by mutation in the gene that codes for the enzyme phenyl alanine hydroxylase (single gene mutation). This manifests itself through phenotypic expression characterised by mental retardation and a reduction in hair and skin pigmentation.

TOPIC CENTRIC EXERCISE -04

Q1. Which of the following illustrates pleiotropy?

(a) In fruit flies, the genes for scarlet eyes and hairy body are located on the same chromosome.
(b) Matings between earless sheep and long-eared sheep always result in short-eared offspring.
(c) Wheat kernels can range from white to red in color trait controlled by several genes.
(d) The human cystic fibrosis gene causes many symptoms, from respiratory distress to digestion problems.

Q2. How many different kinds of gametes will be produced by a plant having the genotype AABbCC?

(a) Two (b) Three
(c) Four (d) Nine

Q3. Inheritance of skin colour in human beings is an example of

(a) Mendelian inheritance
(b) Monogenic inheritance
(c) Complementary genes
(d) Polygenic Quantitative inheritance

Q4. How many gene loci in human controlled skin colour?

(a) Two (b) One
(c) Three (d) Six

Q5. Occasionally, a single gene may express more than one effect. This is

(a) Polygenic inheritance (b) Pleiotropy
(c) Multiple allelism (d) Co-dominance

4.6 SEX DETERMINATION

Establishment of sex through differential development in an individual at an early stage of life, is called sex determination.

There are different methods for sex determination in organisms like environmental, non-allosomic genetic determination, allosomic sex determination and haplodiploidy.

Mechanism of sex determination-

1. **Chromosomal basis of sex determination:**

Chromosomes are of two types:

- Autosomes or somatic chromosomes: These regulate inheritance of somatic characters.
- Sex chromosomes: These chromosomes are associated with sex determination.

Clue Finder

- X- Chromosome discovered by "*Henking*" and called 'X-body'.
- Wilson and Stevens proposed chromosomal theory for sex determination.

(a) Male heterogamety:

In this type male individual produces two different types of gametes. Thus, the sperm determines the sex of the offspring. It involves two types of sex determining mechanisms; XO type and XY type.

(i) XO type (XX-XO type):

It is observed in large number of insects e.g., Grasshopper. Number of chromosomes are different in male and female individuals.

It is clear that, all eggs (ova) bear an additional X-chromosome besides the autosomes while only 50% of the sperms bear X-chromosomes. In grasshopper, eggs fertilised by (A+X) type sperm become females while those fertilised by (A+O) type sperm become males. Therefore, sperm determines the sex of the offspring. Due to the involvement of the X-chromosome in sex determination, it was designated to be the sex chromosome.

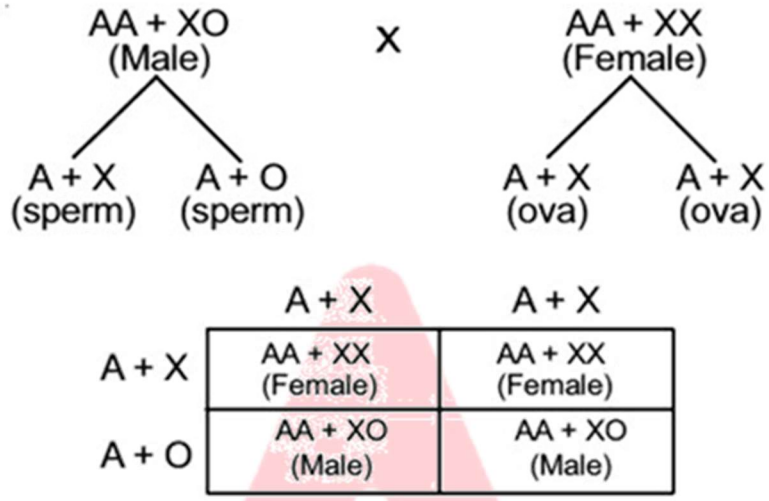


Fig.: XO type of sex determination in Grasshopper

(ii) XX - XY type: This type of sex determination first observed by Wilson & Stevens in an insect.

XX-female and XY-male

In this type of sex determination, female is homogametic, i.e., produces only one type of gamete.

Male is heterogametic, i.e., male produces two types of gametes but both of these gametes have same number of chromosomes.

E.g., *Drosophila, Homo sapiens, Melandrium*

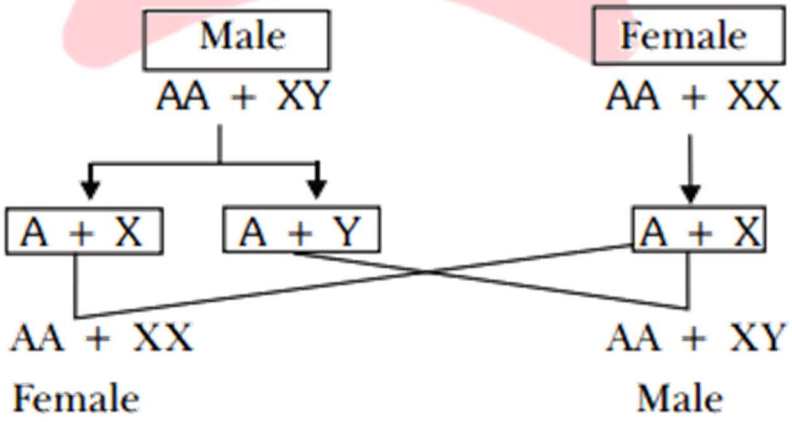


Fig.: XX-XY type of sex determination

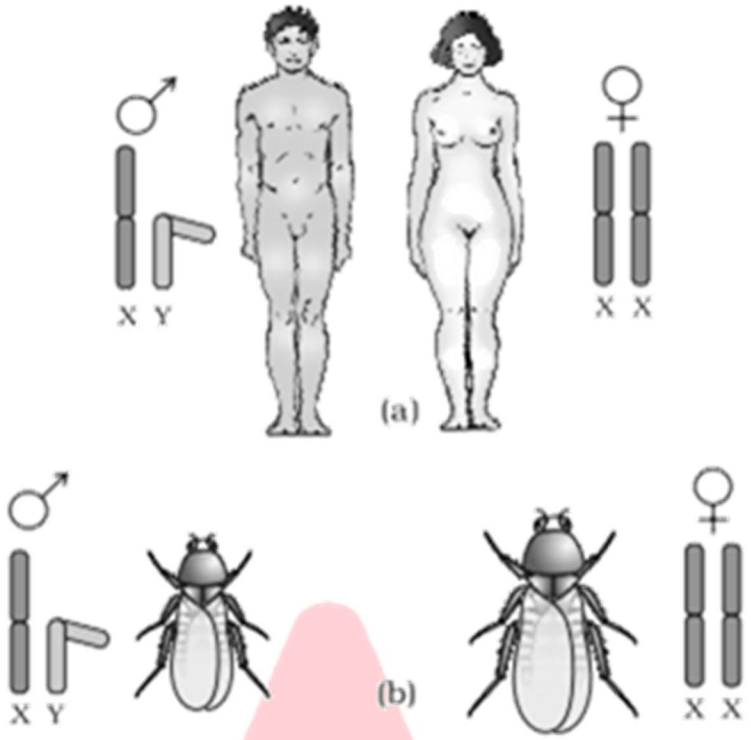


Fig.: Determination of sex by chromosomal differences: (a,b) Both in humans and in Drosophila, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) composition

Critical Thinking

In case the ovum fertilises with a sperm carrying X-chromosome the zygote develops into a female (XX) and the fertilisation of ovum with Y-chromosome carrying sperm results into a male offspring. Thus, it is evident that it is the genetic makeup of the sperm that determines the sex of the child.

(b) Female heterogamety

Female individual produces two different types of gametes. Thus, the egg determines the sex of the offspring. It involves sex-determining mechanisms ZW type:

ZW type (ZW-ZZ type):

In birds, both the sexes possess two sex chromosomes. Unlike human beings, the females contain heteromorphic sex chromosomes while the males have homomorphic sex chromosomes. Because of having heteromorphic sex chromosomes, the females are heterogametic. Different symbols in birds are used to distinguish the female heterogametic in birds (ZW) from male heterogametic sex (XY) in Drosophila and man.

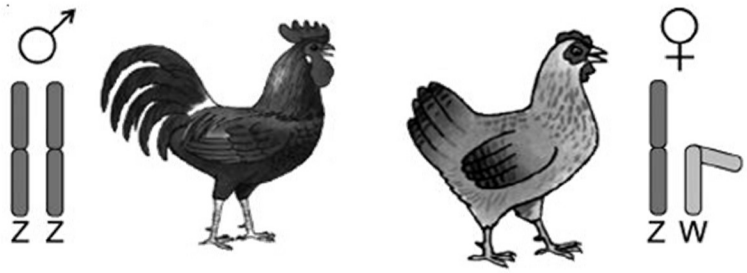


Fig.: In many birds, female has a pair of dissimilar chromosomes ZW and male two similar ZZ chromosomes

4.6.1 Sex Determination in Human Beings

Human beings have 22 pairs of autosomes and one pair of sex chromosomes. All the ova (haploid) formed by female are similar in their chromosome type (22 + X) Therefore females are homogametic. Male individual produces two types of sperms during the process of spermatogenesis. 50% of the total sperm produced possess the X-chromosome and the rest 50% has Y-chromosome besides the autosome. There is an equal probability of fertilisation of the ovum (22 + X) with the sperm carrying either X or Y chromosome. If ovum fertilises with (22 + X) type sperm, the zygote develops into a female (44 + XX) and the fertilisation of ovum with (22 + Y) type sperm results into a male individual (44 + XY) Thus, genetic makeup of the sperm determines the sex of the child. It is also clear that in each pregnancy there is always 50% or 1/2 probability of either a male or a female.

4.6.2 Sex Determination in Honey Bee

The sex determination in honey bee is based on the number of sets of chromosomes an individual receives.
An offspring formed from the union of a sperm and an egg develops as a female (queen or worker), and an unfertilised egg develops as a male (drone) by means of parthenogenesis. This means that the males have half the number of chromosomes than that of a female.

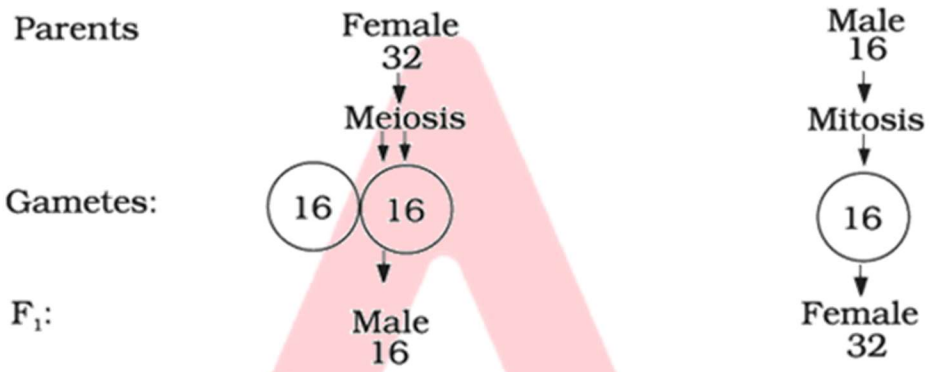


Fig.: Sex Determination in honey bee

The females are diploid having 32 chromosomes and males are haploid, i.e., having 16 choromosomes. This is called as **haplodiploid sex-determination system** and has special characteristic features such as the males produce sperms by mitosis, they do not have father and thus cannot have sons, but have a grandfather and can have grandsons.

TOPIC CENTRIC EXERCISE -05

Q1. In XO type of sex determination
(a) Female produce two different types of gametes
(b) Males produce two different types of gametes
(c) Females produce gametes with Y chromosomes.
(d) Males produce single type of gametes.

Q2. In insects with the XO type of sex determination, what is the genetic makeup of females?
(a) XX (b) XY
(c) XO (d) XXY

Q3. Haplodiploidy is found in
(a) Grasshoppers and cockroaches (b) Birds and reptiles
(c) Butterflies and moths (d) Honeybees, ants and wasps

Q4. An unfertilised egg in honeybees develops as a drone by means of
(a) Parthenogenesis (b) Meiosis
(c) Mitosis (d) Apogamy

Q5. In honeybee, males do not have father because they follow
(a) XO type sex determination (b) XY type sex determination
(c) Haplodiploid sex determination (d) Autosomes based sex determination

4.7 MUTATION

Sudden inheritable change in an organism is called mutation. Mutation results in alternation of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism.

Classification of Mutation

1. Gene Mutation: It is alteration of DNA due to change in nucleotide sequence. Gene mutation may occur due to change in a single base pair of DNA, known as point mutation. A classical example of point mutation is sickle cell anaemia. Change in more than one nucleotide pair is called gross mutation. Gene mutation occurs by following methods.

(a) Frame-shift mutation

Deletion: Removal of one or more bases from nucleotide chain.

Insertion or addition: Addition of one or more bases in a nucleotide chain.

(b) Substitution: The replacement of one base by another. It is of two types:

Transition: When a purine base (A or G) is substituted by another purine base or pyrimidine base (T or C) is substituted by another pyrimidine base.

Transversion: Substitution of a purine base with a pyrimidine base or vice versa.

Gene mutation may occur naturally and automatically due to internal reason. They are named as spontaneous mutations. However, they are produced by external physical or chemical factors. These factors are named as mutagens that are used to create induced mutations.

- **Mutagens**

Mutations can be artificially produced by certain agents called mutagens or mutagenic agents. Following are two major types of mutagens:

1. **Physical mutagens:** Radiations are the most important physical mutagens H.J. Muller who used X-rays, for the first time, to increase the rate of mutation in *Drosophila*, opened an entirely new field in inducing mutations. The main source of spontaneous mutations are the natural radiations coming from cosmic rays of the sun. The spectrum of wavelengths that are shorter (i.e., of higher energy) than the visible light can be subdivided into following two groups:
   (a) Ionizing radiations
   (b) Non-ionizing radiations
   They occur in small amounts in the environment and are known as background radiations.
2. **Chemical mutagens:** Large number of chemical mutagens are now known. These are more injurious than radiations. Chemical mutagens are placed into two groups:
   (a) Those which are mutagenic to both replicating and non-replicating DNA such as nitrous acid.
   (b) Those which are mutagenic only to replicating DNA, such as acridine dyes and base analogues.
   Following are the effects of some of the chemical mutagens:

TOPIC CENTRIC EXERCISE -06

Q1. Which of the following is a classic example of point mutation?
(a) Down's syndrome (b) Haemophilia
(c) Sickle-cell anemia (d) Phenylketonuria

Q2. Mutations result in
(a) Death (b) Change in genetic constitution
(c) Better varieties (d) All above

Q3. Chromosomal aberrations are commonly observed in which type of cells?
(a) Somatic cells (b) Epithelial cells
(c) Cancer cells (d) Cork cells

Q4. Aneuploidy is
(a) Increase in whole number of chromosome
(b) Failure of cytokinesis after telophase stage
(c) Gain or loss of a chromosome
(d) Any of these

Q5. Point mutations are caused by

(a) Substitution (b) Deletion

(c) Insertion (d) All of above

4.8 GENETIC DISORDERS

4.8.1 Pedigree Analysis (Method of study of human genetic disorders):

Human beings, like other living organisms, also follow the principles of inheritance but common Mendelian experiments cannot be carried out over us due to following reasons.

(i) Controlled crosses are not possible in human beings.

(ii) Number of offspring per couple is small.

Because of the reasons described above, human geneticist has to resort to slightly different methods of genetic analysis. Such an analysis of traits in a several of generations of a family is called the pedigree analysis. In human genetics, pedigree study provides a strong tool, which is utilised to trace the inheritance of a specific trait, abnormality or disease. It is useful for the genetic counsellors to advice intending couples about the possibility of having children with genetic defects like haemophilia, colour blindness, phenylketonuria, thalassaemia, sickle cell anaemia (recessive traits), myotonic dystrophy and polydactyly (dominant traits).

A family tree or pedigree is drawn up using certain standard symbols. Some of the important symbols are as follows:

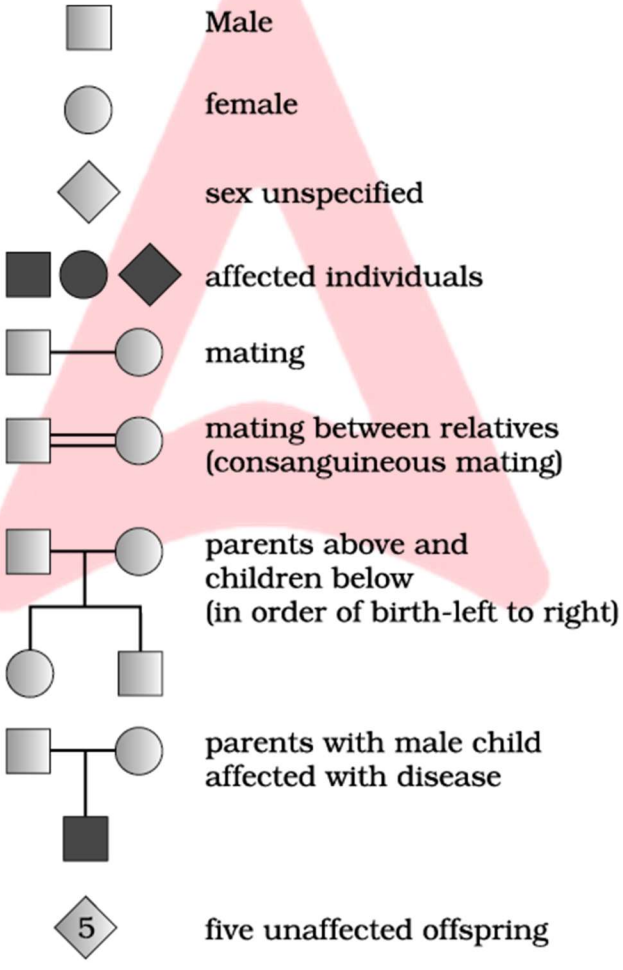


Fig.: Symbols used in the human pedigree analysis

By pedigree analysis one can easily understand whether the trait in question is autosomal dominant or recessive. Similarly, the trait may also be linked to the sex chromosome as in case of haemophilia. For carrying out simple analysis involving recessive or dominant allele, certain clues or simple rules are sought from the pedigree. For example, in the case of recessive allele, characteristic condition can appear in the progeny of apparently unaffected parents. Moreover, two affected individuals cannot have unaffected child. Let us try to understand pedigree analysis with the help of following example.

4.8.2 Mendelian Disorders

These are mainly determined by mutation in the single gene, therefore also called gene related human disorders. They are transmitted to the offspring as per Mendelian principles. The pattern of inheritance of such disorders can be traced in a family by the pedigree analysis. Some common and prevalent Mendelian disorders are as follows:

S.No.	Disorder	Dominant/Recessive	Autosmal/Sex linked
(1)	Haemophilia	Recessive	X-linkled
(2)	Colour blindness	Recessive	X-linkled
(3)	Sickle cell anaemia	Recessive	Autosomal
(4)	Phenylketonuria	Recessive	Autosomal
(5)	Cystic fibrosis	Recessive	Autosomal
(6)	Thalassemia	Recessive	Autosomal
(7)	Myotonic dystrophy	Dominant	Autosomal

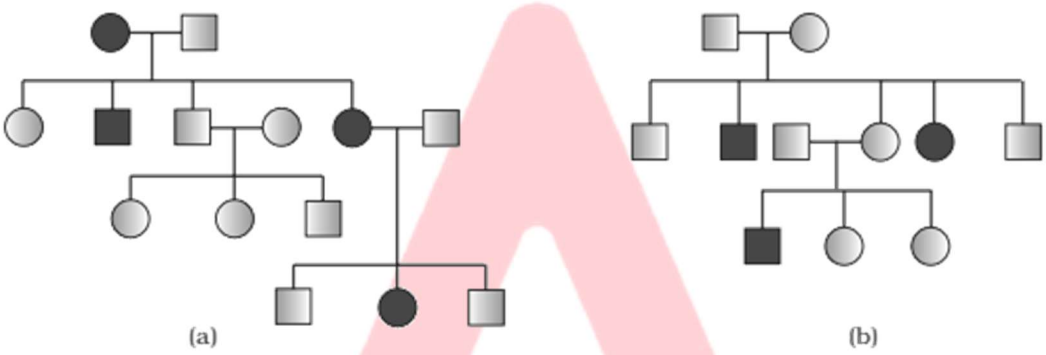


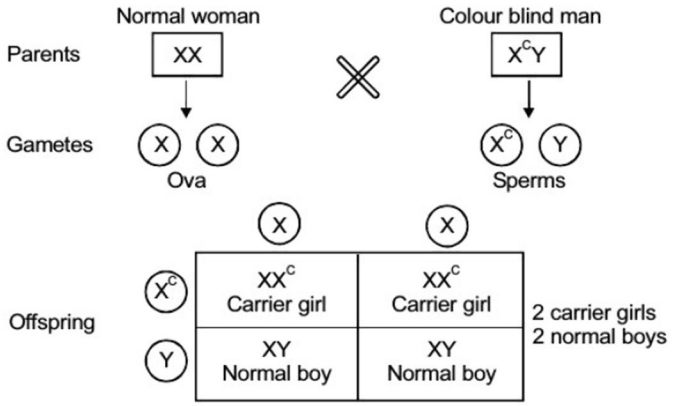
Fig.: Representative pedigree analysis of (a) Autosomal dominant trait (for example: Myotonic dystrophy) (b) Autosomal recessive trait (for example: sickle-cell anaemia)

(i) Colour blindness:

It is a sex-linked (X-linked) recessive disorder due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour. This defect is due to mutation in certain genes present in the X chromosome. It occurs in about 8 per cent of males and only about 0.4 percent of females. This is because the genes that lead to red-green colour blindness are on the X chromosome. Males have only one X chromosome and females have two.

If a colourblind man (X^CY) marries a girl with normal vision (XX), the daughters would have normal vision but would be carrier, while sons would also be normal **(shown in cross(a))**.

Cross (a)



If the carrier girl (heterozygous for colour blindness, X^cX) now marries a colour blind man X^cY, the offspring would show 50% females and 50% males. Of the females, 50% would be carrier for colour blindness and the rest 50% would be colour blind. Of the males, 50% would have normal vision and the 50% would be colour blind **(shown in cross (b))**

Cross (b)

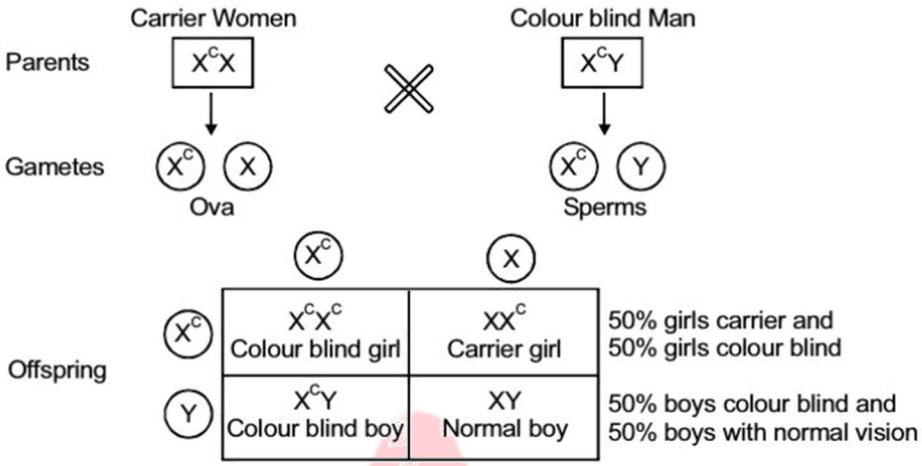


Fig.: Sex-linked inheritance of colour blindness - cross (a) and cross (b)

(ii) Haemophilia:

It is X-linked recessive trait therefore shows its transmission from normal carrier female (heterozygous) to male progeny. Due to presence of defective form of blood clotting factor (protein), exposed blood of affected individuals fails to coagulate.

The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life). Haemophilic female dies before birth. The family pedigree of Queen Victoria shows a number of haemophilic descendents as she was a carrier of the disease.

The person suffering from this disease cannot synthesize a normal blood protein called antihaemophilic globulin (AHG) required for normal blood clotting (Haemophilia A - more severe). Therefore, even a very small cut may lead to continuous bleeding for a long time. This gene is located on X chromosome and is recessive. It remains latent in carrier females.

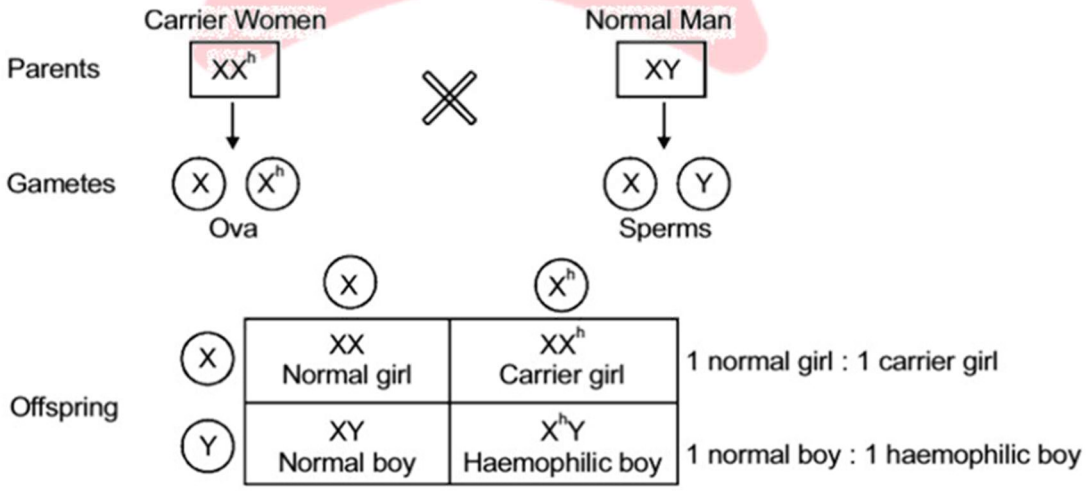


Fig.: Inheritance of haemophilia when the mother is carrier and the father is normal

If a normal man marries a girl who is carrier for haemophilia, the progeny would consist of 50% females and 50% males. Of the females, 50% would be normal and the rest 50% would be hemophilia carrier. Of the males, 50% would be normal and the rest would be haemophiliacs.

(iii) Sickle-cell anaemia:

As it is autosomal recessive disease therefore it can be transmitted from parents to the offspring when both male and female individuals are carrier (heterozygous) for the gene. The disease is controlled by a single pair of allele, Hb^A and Hb^S. Thus three genotypes are possible in population.

(i) Hb^AHb^A (Normal, homozygous)

(ii) Hb^AHb^S (Normal, carrier)

(iii) Hb^SHb^S (Diseased, die before attaining maturity)

Heterozygous ($Hb^A\ Hb^S$) individuals appear apparently unaffected but they are carrier of the disease as there is 50% probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait. The disease/defect is caused by mutation (transversion) of the gene controlling β-chain of haemoglobin. The mutated gene is called Hb^S. Hb^S causes one change in amino acid sequence of β-chain. It replaces glutamic acid (Glu) present at 6th position of the β-chain by amino acid valine (Val). The mutant haemoglobin molecule undergoes polymerisation under low O_2 tension causing the change in the shape of the RBC from biconcave disc to elongated sickle-like structure.

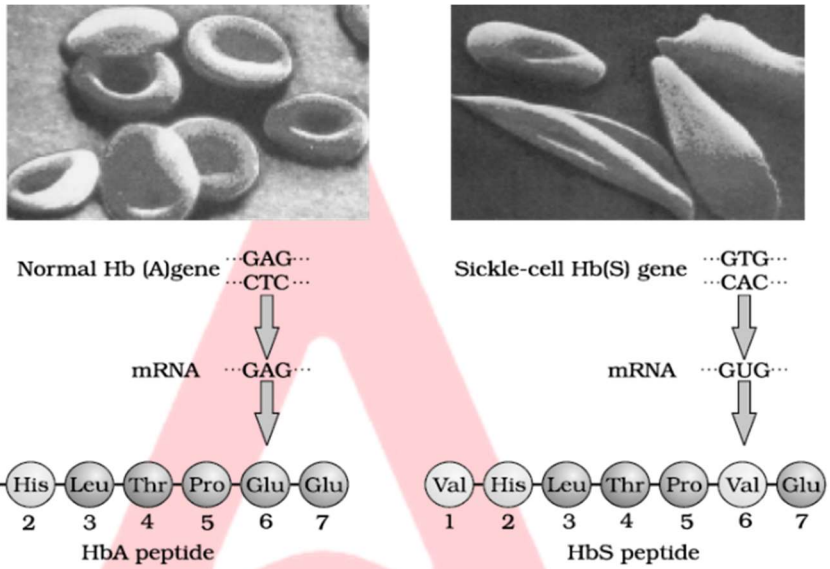


Fig.: Micrograph of the red blood cells and the amino acid composition of the relevant portion of b-chain of haemoglobin: (a) From a normal individual; (b) From an individual with sickle-cell anaemia

(iv) Phenylketonuria:

This inborn error of metabolism is also inherited as the autosomal recessive trait. The affected individual lacks a liver enzyme called phenylalanine hydroxylase that converts the amino acid phenylalanine into tyrosine. As a result of this phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives. Accumulation of these in brain results in mental retardation. These are also excreted through urine because of its poor absorption by kidney.

(v) Thalassemia:

This is also an autosome-linked recessive blood disease transmitted from parents to the offspring when both the partners are unaffected carrier for the gene (or heterozygous). The defect could be due to either mutation or deletion which ultimately results in reduced rate of synthesis of one of the globin chains (α and β chains) that make up haemoglobin. This causes the formation of abnormal haemoglobin molecules resulting into anaemia which is characteristic of the disease. Thalassemia can be classified according to which chain of the haemoglobin molecule is affected.

(a) In α - Thalassemia, production of a globin chain is affected. a Thalassemia is controlled by two closely linked genes HBA1 and HBA2 on chromosome 16 of each parent and it is observed due to mutation or deletion of one or more of the four genes. The more genes affected; the less alpha globin molecules produced.

(b) In β - Thalassemia, production of b globin chain is affected. β- Thalassemia is controlled by a single gene HBB on chromosome 11 of each parent and occurs due to mutation of one or both the genes.

Thalassemia differs from sickle-cell anaemia in that the former is a quantitative problem of synthesising too few globin molecules while the latter is a qualitative problem of synthesising an incorrectly functioning globin.

4.8.3 Chromosomal Disorders:

The chromosomal disorders on the other hand are caused due to absence or excess or abnormal arrangement of one or more chromosomes. It is caused due to absence or excess or abnormal arrangement of one or more chromosome. Failure of segregation of chromatids/Non-disjunction of chromosomes during cell division cycle results in the gain or loss of chromosome(s), called **Aneuploidy**. Down's syndrome, Klinefelter's syndrome and Turner's syndrome are common examples of chromosomal disorders. Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosome in an organism and this phenomenon is called **polyploidy**. This condition is often seen in plants.

(a) Down's syndrome: It was first described in 1866 by Langdon Down. The disorder develops due to trisomy of chromosome number 21. Trisomic condition arises due to the formation of n+1 male or female gamete by non-disjunction and the subsequent fertilisation by a normal (n) gamete. It is characterised by:

(i) Short stature
(ii) Small round head
(iii) Furrowed tongue
(iv) Partially open mouth
(v) Broad palm with characteristic palm crease
(vi) Many 'loops' on finger tips
(vii) Big and wrinkled tongue
(viii) Physical (underdeveloped gonads and genitals, loose jointed-ness), psychomotor and mental development is retarded.

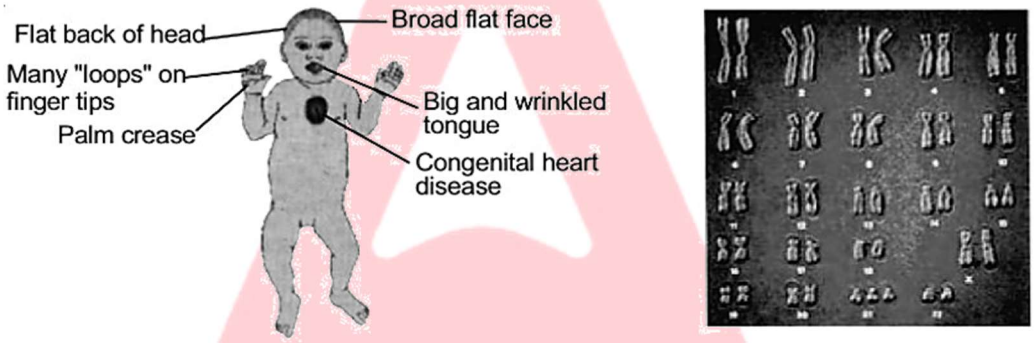


Fig.: A representative figure showing an individual inflicted with Down's syndrome

(b) Klinefelter's syndrome: It is caused due to the presence of an additional copy of X-chromosome (b resulting into 44 + XXY type **chromosome** complement. The defect appears due to union of an abnormal egg (22 + XX) and a normal sperm (22 + Y) or normal egg (22 + X) and abnormal sperm (22 + XY) Such persons are sterile males with overall masculine development and some female characteristics (e.g., Feminine pitched voice, development of breast or gynaecomastia).

(c) Turner's syndrome: The disorder is due to monosomy. It appears due to fusion of abnormal egg (22+ 0) and a normal sperm (22 + X) or a normal **egg** (22 + X) and abnormal sperm (22 + 0). Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.

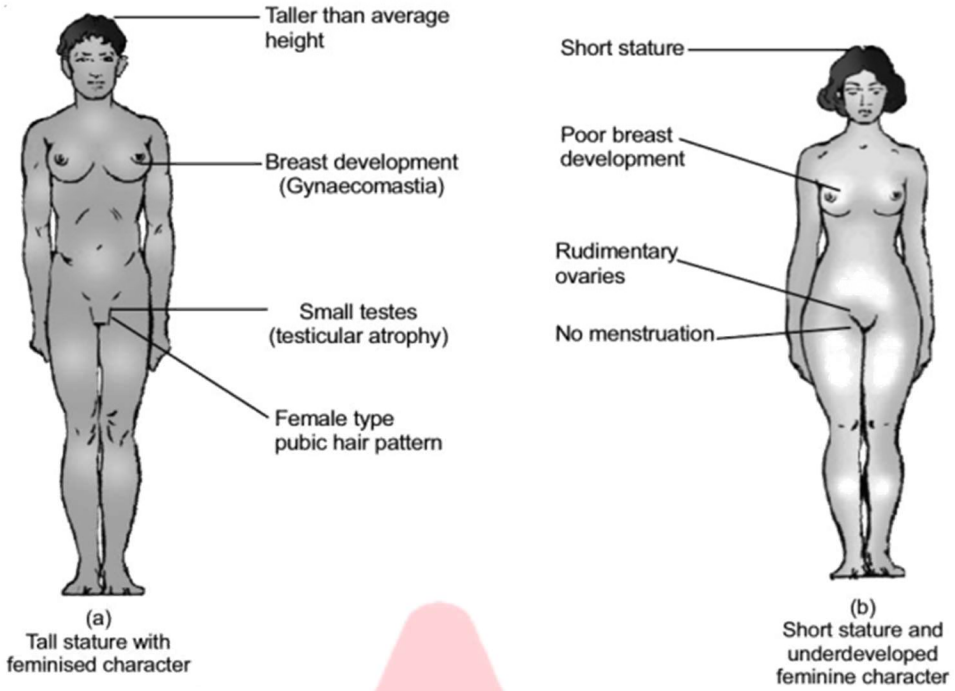


Fig.: Representative figures showing individuals inflicted with (a) Klinefelter syndrome and (b) Turner's syndrome

TOPIC CENTRIC EXERCISE -07

Q1. **Sickle cell anemia is caused because of**
(a) Substitution of glutamic acid by valine at 6th position of β- globin chain
(b) Substitution of valine by glutamic acid at 6th position of β- globulin chain
(c) Substitution of glutamic acid by valine at 6th position α- globin chain.
(d) Substitution of valine by glutamic acid at 6th position of α- globin chain

Q2. **Down syndrome is**
(a) Failure of segregation of chromosome
(b) Failure of telophase
(c) Presence of addition copy of chromosome number 21
(d) Presence of addition copy of chromosome number 17

Q3. **A woman with normal vision, but whose father was colour blind, marries a colour blind man. Suppose that the fourth child of this couple was a boy. This boy-**
(a) Must have normal colour vision
(b) May be colour blind or may be normal vision
(c) Will be partially colour blind since he is heterozygous for the colour blind mutant allele
(d) Must be colour blind

Q4. **Rudimentary ovary and lack of secondary sexual characters are symptoms of**
(a) Down syndrome (b) Phenylketonuria
(c) Klinefelter's syndrome (d) Turner's syndrome

Q5. **What is the main cause of Thalassemia?**
(a) Overproduction of haemoglobin molecules
(b) Mutation or deletion affecting the synthesis of globin chains
(c) Abnormal functioning of globin chains
(d) Mutation in the HBB gene

Solved Examples

Ex: 1. **In his classic experiments on pea plants, Mendel did not use:**
(a) Seed colour (b) Pod length

(c) Seed shape (d) Flower position

Sol. **(b)** Seven pairs of contrasting characters in pea plants were studied by Mendel in his experiments. They were: i) form of seed; ii) colour of cotyledons; iii) colour of seed coat; iv) form of the pod; v) colour of the pod; vi) position of flower; vii) height of plant (length of the stem).

Ex: 2. **Thalassemia and sickle cell anemia is caused due to a problem in globin molecule synthesis. Select the correct statement**

(a) Both are due to a qualitative defect in globin chain synthesis
(b) Both are due to a quantitative defect in globin chain synthesis
(c) Thalassemia is due to less synthesis of globin molecules
(d) Sickle cell anemia is due to a quantitative problem of globin molecules

Sol. **(c)** Thalassemia and sickle cell anemia are genetic disorders in which hemoglobin synthesis is affected. In thalassemia, there is a defect in the a or ß globin chain. This leads to the synthesis of abnormal red blood cells due to the decreased synthesis of globin molecules.

Ex: 3. **If one parent has blood group A and the other parent has blood group B, the offspring have which blood group?**

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

Sol. **(c)** If one parent has blood group A (genotype IAIA or IAi) and the other has blood group B (genotype IBIB or IBi), the possible genotypes of the offspring can be: I^AI^B (Blood group AB), I^Ai (Blood group A), I^Bi (Blood group B), ii (Blood group O)

Ex: 4. **An abnormal human baby with 'XXX' sex chromosomes was born due to**

(a) Formation of abnormal sperms in the father
(b) Formation of abnormal ova in the mother
(c) A fusion of two ova and one sperm
(d) Fusion of two sperms and one ovum

Sol. **(b)** The XXX condition, also known as Triple X syndrome, typically occurs due to nondisjunction during meiosis in the mother, resulting in an ovum with two X chromosomes instead of one. When fertilized by a normal X-carrying sperm, it leads to an XXX genotype

Ex: 5. **A human female with Turner's syndrome**

(a) Has 45 chromosomes with XO
(b) Has one additional X chromosomes
(c) Exhibits male characters
(d) Is able to produce children with a normal husband

Sol. **(a)** Turner syndrome, a condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety medical and developmental problems, including short height, failure of the ovaries to develop and heart defects.

Ex: 6. **Which Mendelian idea is depicted by a cross in which the F1 generation resembles both the parents?**

(a) Incomplete dominance (b) Law of dominance
(c) Inheritance of one gene (d) Codominance

Sol. **(d)** In codominance, a heterozygous individual expresses both simultaneously without any blending

Ex: 7. **The incorrect statement with regards to hemophilia is**

(a) It is a sex-linked disease
(b) It is a recessive disease
(c) It is a dominant disease
(d) A single protein involved in the clotting of blood effected

Sol. **(c)** Hemophilia is a sex-linked and recessive disorder and not dominant.

Ex: 8. **If both parents are carriers for thalassemia, which is an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child?**

(a) No chance (b) 50%

(c) 25% (d) 100%

Sol. **(c)** When two carriers have a child, there is a 25% chance the child will inherit two copies of the mutated gene (be affected), a 50% chance of being a carrier (having one normal and one mutated gene), and a 25% chance of inheriting two normal genes (unaffected and not a carrier).

Ex: 9. **F_2 generation in a Mendelian cross showed that both genotypic and phenotypic ratios are the same as 1:2:1. It represents a case of**

(a) Codominance (b) Dihybrid cross

(c) Monohybrid cross with complete (d) Incomplete dominance

Sol. **(d)** In incomplete dominance, the heterozygous genotype expresses a phenotype that is intermediate between the two homozygous phenotypes. As a result, the F_2 generation shows a genotypic and phenotypic ratio of 1:2:1

Ex: 10. **Which one of the following conditions in humans is correctly matched with its chromosomal abnormality/linkage?**

(a) Klinefelter's syndrome - 44 autosomes + XXY

(b) Colour blindness - Y-linked

(c) Erythroblastosis fetalis - X-linked

(d) Down syndrome - 44 autosomes + XO

Sol. **(a)** Klinefelter's syndrome is a genetic disorder caused by the presence of an extra X chromosome in males, resulting in the karyotype 44 autosomes + XXY

Exercise-01 Level -01

1. F_2 generation is produced by
(a) Crossing F_1 progeny with one of the parents
(b) Selfing the F_1 progeny of two individual parents
(c) Selfing the parents
(d) Recessive cross between individual parents.

2. An example of codominance is
(a) Eye colour in *Drosophila*
(b) Seed shape and colour in pea
(c) AB blood group in man
(d) Haemophilia in man.

3. Multiple alleles control inheritance of
(a) Colour blindness
(b) Sickle cell anaemia
(c) Blood group
(d) Phenylketonuria

4. A child with mother of A blood group and father of AB blood group will not have the following blood group.
(a) A (b) B
(c) AB (d) O

5. Select the pair of dominant traits studied by Mendel.
(a) Axial flower, green pod, green seed
(b) Green pod, inflated pod, axial flower
(c) Yellow seed, violet flower, yellow pod
(d) Round seed, constricted pod, axial flower

6. Select the incorrect statement regarding linkage.
(a) Complete linkage produces parental types and no recombinants in progeny.
(b) There is no crossing over between linked genes.
(c) Incompletely linked genes are located very closely in the same chromosome and are inherited together over the generations.
(d) Completely linked genes do not segregate independently.

7. If F_1 generation has all tall plants and ratio of F_2 generation is 3 tall: 1 dwarf, it proves
(a) Law of independent assortment
(b) Law of segregation
(c) Law of codominance
(d) Incomplete dominance

8. Inheritance of ABO blood groups is an example of
(i) Incomplete dominance
(ii) Dominance
(iii) Codominance
(iv) Multiple allelism
(a) (i) and (iii) only (b) (ii) and (iii) only
(c) (ii), (iii) and (iv) (d) (i), (iii) and (iv)

9. In a plant, red fruit (R) is dominant over yellow fruit (r) and tallness (T) is dominant over shortness (t). A plant with RRTt genotype is crossed with plant of rrtt genotype. The progeny is
(a) all red fruit, 25% tall
(b) all red fruit, 50% tall
(c) all red fruit, 75% tall
(d) all red fruit and tall

10. During meiosis all alleles of a genomic pair segregate from each other. How many alleles are passed onto a gamete?
(a) One (b) Two
(c) Four (d) Six

11. A human male produces diallelic sperms in equal proportion AB, Ab, aB and ab. The genotype of the person would be
(a) AaBb (b) AaBB
(c) AABb (d) AABB

12. In garden pea, the gene which controls the flower colour also controls the colour of seed and presence of red spots in leaf axils. It is an example of
(a) Polygenic inheritance
(b) Co-dominance
(c) Multiple allelism
(d) Pleiotropism

13. Which of the following pair of features is a good example of polygenic inheritance?
(a) Human height and skin colour
(b) ABO blood group in humans and flower colour of *Antirrhinum*.
(c) Hair pigment of mouse and tongue rolling in humans.
(d) Human hair colour and sickle cell anaemia

14. Which one of the following is an example of polygenic inheritance
(a) Skin colour in humans
(b) Flower colour in *Antirrhinum*
(c) Production of male honey bee
(d) Pod shape in garden pea

15. Which statement is incorrect for polygenic inheritance?
(a) Polygenic traits are controlled by three or more genes.
(b) Environment has lesser effect on phenotypic expression.

(c) It controls quantitative traits.
(d) Both (b) and (c)

16. A mutation is a
(a) Sudden temporary change in an organism's genetic material
(b) Spontaneous change that occurs in response to specific external factors and chemicals
(c) Change in hereditary material directed by a changing environment
(d) Change in genotype which may result in a new expression of a characteristic.

17. Which is incorrect about colour blindness?
(a) This is due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour.
(b) A daughter will not normally be colourblind, unless her mother is a carrier and her father is colourblind.
(c) If female has X^CX, then it is called carrier but when male has X^CY then it will be colourblind.
(d) Out of total population of son of carrier woman, 25% have chances of being colourblind.

18. In human sperm, beside autosome the chromosome complement contains
(a) X and Y (b) Either X or Y
(c) Y only (d) X only

19. A disease which is inherited as an autosomal recessive condition is
(i) Haemophilia
(ii) Sickle cell anaemia
(iii) Colour blindness
(iv) Thalassemia.
(a) (i), (iii) and (iv) (b) (ii) and (iv)
(c) (i) and (iii) (d) (ii) and (iii)

20. A person with chromosome complement of 47, XXY suffers from
(a) Klinefelter's syndrome
(b) Down's syndrome
(c) Turner's syndrome
(d) sickle cell anaemia

21. Sex chromosomes of female is heteromorphic and male is homomorphic in
(a) Human beings (b) Birds
(c) Honey bees (d) All of these

22. Male contains half of the chromosomes than that present in female in
(a) Human beings (b) Pigeon
(c) Honey bee (d) Moth

23. How many chromosomes are present in each cell of a person suffering from Down's syndrome?
(a) 21 (b) 23
(c) 45 (d) 47

24. Which amino acid is substituted in sickle cell anaemia?
(a) Glutamic acid by valine in α-chain
(b) Glutamic acid by valine in β-chain
(c) Valine by glutamic acid in β-chain
(d) Valine by glutamic acid in α-chain

25. A man and a woman who do not show any apparent signs of a certain inherited disease, have five children (2 daughters and 3 sons). All sons suffer from the given disease but none of the daughters were affected. Which of the following could be this disease?
(a) Autosomal recessive diseases
(b) Sex-linked recessive disease
(c) Sex-linked dominant
(d) Autosomal dominant disease

26. Select the incorrect statement.
(a) Human males have one of their sex-chromosome much shorter than other.
(b) In birds, females show heterogamety.
(c) In male grasshoppers, 50% of sperms have no sex-chromosome.
(d) None of these

27. Phenylketonuria disorder is due to
(a) Recessive autosomal genes
(b) Dominant autosomal genes
(c) Dominant sex genes
(d) Recessive sex genes.

28. In which disease, the RBC of a person changes from biconcave to elongated structure?
(a) Haemophilia (b) Sickle-cell anaemia
(c) Thalasssemia (d) Leukemia

29. How many contrasting traits of pea pod were chosen by Mendel?
(a) 7 (b) 2
(c) 4 (d) 3

30. Heterozygous tall plant is selfed. It produces both tall and dwarf plants. This confirms Mendel's
(a) law of dominance
(b) law of segregation
(c) law of independent assortments
(d) incomplete dominance.

31. A child with O blood group has father with blood group B, then what will be the genotype of father?
(a) I^OI^O (b) I^BI^B
(c) I^AI^B (d) I^BI^O

32. "Gametes are never hybrid. It is a statement of law of
(a) Dominance
(b) Segregation
(c) Independent assortment

(d) Random fertilisation

33. Who used frequency of recombination between gene pairs on the same chromosome as a measure of distance between genes and mapped their position on chromosome?
(a) Alfred Sturtevant (b) Gregor Mendel
(c) Correns (d) Tschermak

34. Number of genotypes found in F_2 progeny of a dihybrid cross is
(a) 9 (b) 6
(c) 3 (d) 1

35. In a dihybrid cross AABB × aabb, F_2 progeny of AABB, AABb, AaBB and AaBb occurs in the ratio of
(a) 1:1:1:1 (b) 9:3:3:1
(c) 1:2:2:1 (d) 1:2:2:4

36. AB genes are linked. What is genotype of the progeny in a cross between AB/ab and ab/ab?
(a) AABB and aabb (b) AaBb and aabb
(c) AAbb and aaBB (d) AaBb and AaBb

37. Blood grouping in human beings is controlled by
(a) 4 alleles in which A is dominant
(b) 3 alleles in which A and B are codominant and i is recessive
(c) 3 alleles in which none is dominant
(d) 3 alleles in which only one is dominant.

38. In a certain taxon of insects some have 32 chromosomes and the others have 31 chromosomes. The 32 and 31 chromosome-bearing organisms are
(a) Males and females, respectively
(b) Females and males, respectively
(c) Drones and males, respectively
(d) Males and drones, respectively.

39. Experimental verification of chromosomal theory of inheritance was given by
(a) Thomas Hunt Morgan
(b) Gregor Johann Mendel
(c) Hugo de Vries
(d) Langdon Down

40. Genotype is
(a) Genetic composition of many organisms
(b) Genetic composition of plastids
(c) Genetic composition of germ cells
(d) Genetic composition of an individual.

41. Sex determination in grasshoppers, humans and Drosophila is similar because
(a) Females are hemizygous
(b) Males have one X-chromosome and females have two X-chromosomes
(c) All males always have one Y-chromosome in all three species
(d) The ratio of autosomes to sex chromosomes is the same in all three organisms.

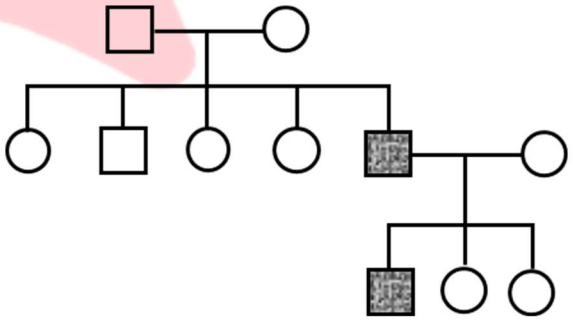
42. If there is complete linkage in F_2 generation
(a) Parental types and recombinants appear in equal ratio
(b) Recombinants are less than parental types
(c) Recombinants are more than parental types
(d) There will be only parental types

43. Select the incorrect statement.
(a) A gamete contains only one of the two alleles of a character.
(b) In diploid cells, genes occur in allelic pairs.
(c) T.H. Morgan worked with tiny *Drosophila melanogaster*
(d) Linked genes show dihybrid ratio of 9:3:3:1

44. Some of the dominant traits studied by Mendel were
(a) Round seed shape, green seed colour, and axial flower position
(b) Terminal flower position, green pod colour and inflated pod shape
(c) Violet flower colour, green pod colour, and round seed shape
(d) Wrinkled seed shape, yellow pod colour, and axial flower position.

45. A colourblind man (X^cY) has a colourblind sister (X^cX^c) and a normal brother (XY). What is genotype of father and mother respectively?
(a) X^cY, X^cX^c (b) XY, X^cX
(c) XY, X^cX^c (d) X^cY, X^cX

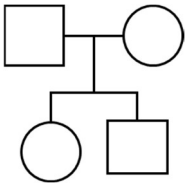
46. In the following pedigree chart, the mutant trait is shaded. The gene responsible for the trait is

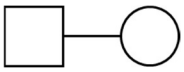


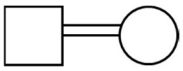
(a) Dominant and sex linked
(b) Dominant and autosomal
(c) Recessive and sex linked
(d) Recessive and autosomal

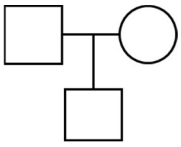
47. Which of the following individuals are affected by sickle cell anaemia?
(a) Hb^SHb^A (b) Hb^SHb^S
(c) Hb^AHb^A (d) Both (a) and (b)

48. Which of the following symbol represents mating between two

(a) 

(b) 

(c) 

(d) 

49. In snapdragon plant, the gene for red flower colour (R) is incompletely dominant over the gene for white flower colour (r), hence the plants heterozygous for flower colour have pink flowers (Rr). What will be ratio of offsprings in a cross between two pink flowered plant?
(a) 75% red flowers, 25% pink flowers
(b) All red flowers
(c) 50% red flowers, 50% pink flowers
(d) Red: Pink: White: 1:2:1

50. A gene showing codominance has
(a) Alleles tightly linked on the same chromosome
(b) Alleles that are recessive to each other
(c) Both alleles independently expressed in the heterozygote
(d) One allele dominant on the other.

Exercise-02 Level -02

1. Select the correct statement regarding the flower colour in pea plant.
(a) White flower colour is a dominant trait.
(b) Violet flower colour is a recessive trait.
(c) Yellow flower colour is a dominant trait.
(d) White flower colour is a recessive trait.

2. The percentage of heterozygous progenies obtained in F_2 generation in a cross between pure tall and dwarf parents is-
(a) 50 % (b) 20 %
(c) 25 % (d) 100 %

3. Though F_1 generation shows all tall plants, and F_2 generation witness dwarf plants also. It proves
(a) law of dominance
(b) independent assortment
(c) law of segregation
(d) linkage

4. Read the following and identify the wrong statement.
(a) During Mendel's investigation, statistical analysis and mathematical logic were applied to problems of biology for the first time.
(b) Mendel selected 14 true breeding pea varieties.
(c) Mendel was the first to conduct hybridisation experiments.
(d) Mendel's experiments had large sampling size which gave grater credibility to the data collected.

5. If in a dihybrid cross Mendel had used two such characters which have linked, he would have faced difficulty in explaining the results on the basis of his
(a) Law of segregation
(b) Law of multiple factor hypothesis
(c) Law of independent assortment
(d) Law of dominance

6. In Mendel's experiments with garden pea, round seed shape (RR) was dominant over wrinkled seeds (rr), yellow cotyledon (YY) was dominant over green cotyledon (yy). What are the expected phenotypes in the F_2 generation of the cross RRYY × rryy?
(a) Only wrinkled seeds with green cotyledons
(b) Round seeds with yellow cotyledons and wrinkled seeds with yellow cotyledons
(c) Only round seeds with green cotyledons
(d) Only wrinkled seeds with yellow cotyledons

7. Which of the following statements is not true for the two genes that show 50% recombination frequency?
(a) If the genes are present on the same chromosome, they undergo more than one cross overs in every meiosis.
(b) The genes may be on different chromosomes.
(c) The genes are tightly linked.
(d) The genes show independent assortment,

8. In XO type of sex determination
(a) Females produce two different types of gametes

(b) Males produce two different types of gametes
(c) Females produce gametes with Y chromosomes
(d) Males produce single type of gametes

9. Which one of the following conditions of the zygotic cell would lead to the birth of a normal human female child?
(a) Two X chromosomes
(b) Only one Y chromosome
(c) Only one X chromosome
(d) One X and one Y chromosome

10. Identify the wrong statement.
(a) In male grasshoppers, 50% of the sperms have no sex chromosome
(b) Usually female birds produce two types of gametes based on sex chromosomes.
(c) The human males have one of their sex chromosomes much shorter than the other.
(d) The male fruit fly is homogametic.

11. In a dihybrid cross where two parents differ in two pairs of contrasting traits like seed colour yellow (YY) and seed colour green (yy) with seed shape round (RR) and seed shape wrinkled (rr), the number of green coloured seeds (yy) among sixteen products of F_2 generation when homozygous dominant is crossed with homozygous recessive will be
(a) 2 (b) 4
(c) 6 (d) 8

12. A woman having genes for haemophilia on one X chromosome and other gene for colourblindness on another X chromosomes marries a normal man. The progeny will show
(a) 50% haemophilic colour-blind sons and 50% normal sons.
(b) 50% daughters are carrier for each haemophilia and colour-blindness
(c) All sons and daughters are haemophilic and colour-blind.
(d) Haemophilic and colour-blind daughters.

13. Sickle-cell anaemia is
(a) caused by substitution of valine by glutamic acid in the beta globin chain of haemoglobin
(b) caused by a change in a single base pair of DNA
(c) single base substitution at the fifth codon of alpha globin gene
(d) an autosomal linked dominant trait

14. Both husband and wife have normal vision though their fathers were colourblind. The probability of their daughter becoming colourblind is
(a) 0% (b) 25%
(c) 50% (d) 75%

15. Haemophilia is more common in males because it is a
(a) recessive character carried by Y-chromosome
(b) dominant character carried by Y-chromosome
(c) dominant trait carried by X-chromosome
(d) recessive trait carried by X-chromosome

16. Pick out the correct statements.
A. Haemophilia is a sex-linked recessive disease.
B. Down's syndrome is characterised by palm crease.
C. Phenylketonuria is an autosomal recessive trait.
D. Sickle cell anaemia is an X-linked recessive trait.
(a) (A) and (D) are correct.
(b) (B) and (D) are correct.
(c) (A), (C) and (D) are correct.
(d) (A), (B) and (C) are correct.

17. If both parents are carriers for thalessemia, which in an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child?
(a) 100% (b) No chance
(c) 50% (d) 25%

18. Incomplete dominance is different from codominance as in former
(a) There is no mixing of effect
(b) The hybrid phenotype is an intermediate blend of the two parent phenotypes
(c) Hybrid possesses same phenotype that of parents
(d) A quantitative effect is absent

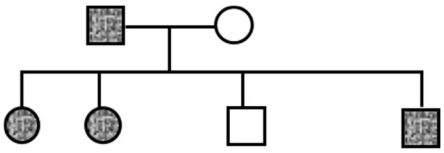
19. Select the incorrect statement.
(a) Chromosomes as well as genes occur in pairs.
(b) Mendel could not provide any physical proof for the existence of factors.
(c) In the Mendelian dihybrid cross, segregation of one pair of characters is independent from the segregation of the other pair of characters.
(d) Distantly located genes assort together.

20. Which pair of features represents polygenic inheritance?
(a) Human eye colour and sickle cell anaemia.
(b) Hair pigment of mouse and tongue rolling in humans.
(c) ABO blood groups in humans and flower colour of *Mirabilis jalapa*.
(d) Human height and skin colour

21. Select the correct statement.
(a) Tightly linked genes on the same chromosome show very few recombination.
(b) Tightly linked genes on the same chromosome show higher recombination.

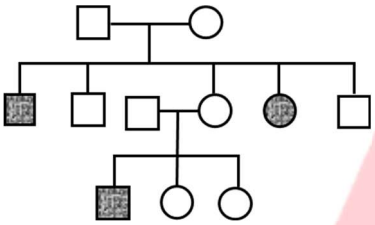
(c) Genes apart on the same chromosome show very few recombination.
(d) Genes loosely linked on the same chromosome show similar recombination as the tightly linked ones.

22. Study the pedigree chart of a certain family given below and select the correct conclusion which can be drawn for the character.



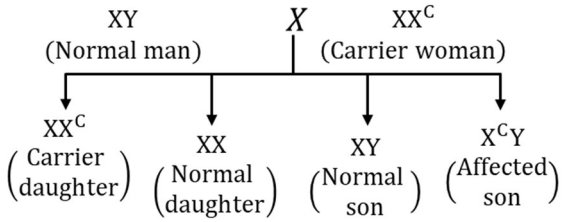
(a) The female parent is heterozygous.
(b) The parents could not have had a normal daughter for this character.
(c) The trait under study could not be colour blind.
(d) The male parent is homozygous dominant.

23. Study the pedigree chart given below. What does it show?



(a) Inheritance of a condition like phenylketonuria as an autosomal recessive trait.
(b) The pedigree chart is wrong as this is not possible.
(c) Inheritance of a recessive sex-linked disease like haemophilia.
(d) Inheritance of a sex-linked inborn error of metabolism like myotonic dystrophy.

24. Inheritance of which of the following traits is shown in the given cross?



(a) X-linked dominant trait
(b) X-linked recessive trait
(c) Autosomal recessive trait
(d) Autosomal dominant trait

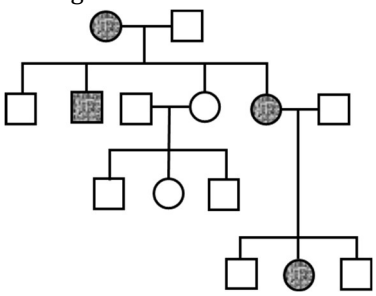
25. What would be the number of chromosomes in the ovum (fertilised by a normal sperm) that resulted in the appearance of Klinefelter's syndrome in the offspring?

(a) 23 (b) 22
(c) 21 (d) 24

26. The number of phenotype recombinant offsprings formed during F_2 generation of a dihybrid cross is

(a) 9/16 (b) 7/16
(c) 6/16 (d) 4/16

27. Find out the correct pattern of inheritance in the pedigree chart given below.



(a) X-linked dominant trait
(b) X-linked recessive trait
(c) Autosomal dominant trait
(d) Autosomal recessive trait

28. Which one is the incorrect statement with regards to the importance of pedigree analysis?

(a) It confirms that DNA is the carrier of genetic information.
(b) It helps to understand whether the trait in question is dominant or recessive.
(c) It confirms whether the trait is linked to one of the autosomes.
(d) It helps to trace the inheritance of a specific trait.

29. If a boy's father has haemophilia and his mother has one gene for haemophilia, what is the chance that the boy will inherit the disease?

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

30. How many genes control β thalassemia?

(a) Two (b) Three
(c) Four (d) One

31. Pure red flowered plants are crossed with pure white flowered plants in an example of incomplete dominance, and the F_1 individuals have all pink flowers. Which one of the following is not correct?

(a) If self-pollinated, pink flowered plant will produce only pink flowered offsprings
(b) If self-pollinated, pink flowered plant will produce offsprings having three kinds of flowers.
(c) If self-pollinated, the genes of the hybrid pink flowered plant will segregate
(d) If self-pollinated, half of the offsprings of the pink flowered plant will be homozygous

32. A tall true breeding garden pea plant is crossed with a dwarf true breeding garden pea plant. When the F_1 plant were selfed the resulting genotypes were in the ratio of
(a) 1:2:1: Tall homozygous: Tall heterozygous: Dwarf
(b) 1:2:1: Tall heterozygous: Tall homozygous: Dwarf
(c) 3:1: Tall: Dwarf
(d) 3:1: Dwarf: Tall

33. What is the genotypic and phenotypic ratio of monohybrid test cross?
(a) 1:1 (b) 1:2
(c) 3:1 (d) 1:2:1

34. Appearance of hidden character in some progeny of F_2 population indicates
(a) Law of purity of gametes
(b) Law of independent assortment
(c) Law of dominance
(d) Complementary gene

35. With respect to Down's syndrome, identify the incorrect statement.
(a) It is due to trisomy of 11.
(b) Affected individual has furrowed tongue and partially open mouth.
(c) Palm is broad with characteristic palm crease.
(d) Mental development is retarded.

36. In which of the following syndrome gynaecomastia is expressed?
(a) Klinefelter's syndrome
(b) Turner's syndrome
(c) Down's syndrome
(d) Both (b) and (c)

37. A boy is colourblind, out of his two sisters one is colourblind and one is carrier (normal) then who is colourblind in his family?
(a) Father
(b) His grandfather and mother
(c) Mother
(d) His grandfather

38. Select incorrect statement with regard to haemophilia.
(a) A single protein involved in the clotting of blood is affected.
(b) It is a sex-linked disease.
(c) It is a recessive disease
(d) It is a dominant disease

39. In which of the following diseases, the man has an extra X-chromosome?
(a) Turner's syndrome
(b) Klinefelter's syndrome
(c) Down's syndrome
(d) Haemophilia

40. Independent assortment of genes does not take place when
(a) Genes are located on homologous chromosomes
(b) Genes are linked and located on same chromosomes
(c) Genes are located on non-homologous chromosomes
(d) All of these

41. Pleiotropy is a condition in which a single gene
(a) Controls only one phenotype
(b) Controls more than one phenotype
(c) Does not control any phenotype
(d) None of these

42. Mendel's Law of independent assortment best suited for genes situated on the
(a) Non-homologous chromosomes
(b) Homologous chromosomes
(c) Extra nuclear genetic element
(d) Same chromosome

43. Which one of the following cannot be explained on the basis of Mendel's law of dominance?
(a) The discrete unit controlling a particular character is called a factor
(b) Out of one pair of factors one is dominant, and the other recessive.
(c) Alleles do not show any blending and both the characters recover as such in F_2 generation.
(d) Factors occur in pairs.

44. Mendel self-pollinated the F_2 recessive plants. The offspring produced will have
(a) 50% recessive traits, 50% dominant traits
(b) 75% dominant traits, 25% recessive traits
(c) 100% dominant traits
(d) 100% recessive traits.

45. The genotypes of a husband and wife are I^AI^B and I^A. Among the blood types of their children, how many different genotypes and phenotypes are possible?
(a) 3 genotypes, 4 phenotypes
(b) 4 genotypes, 3 phenotypes
(c) 4 genotypes, 4 phenotypes
(d) 3 genotypes, 3 phenotypes

46. In pea hybrids obtained from red flowered and white flowered strains were crossed back to pure red flowered strain. The progeny of this cross will have
(a) Red flowers only
(b) White flowers only
(c) Equal number of red and white flowers
(d) Mostly white flowers but few red flowers.

47. A true breeding plant is
(a) One that undergone continuous self-pollination
(b) Always homozygous recessive in its genetic constitution
(c) Produced due to cross-pollination among unrelated plants
(d) None of these

48. The colour based contrasting traits out of seven contrasting pairs,
(a) 1 (b) 2
(c) 3 (d) 4

49. Thalassemia and sickle cell anaemia are similar as
(a) Both are quantitative problem of synthesizing too few globin molecules
(b) Both are qualitative problem of synthesizing incorrectly functioning globin protein.
(c) These are X linked disorders
(d) These are autosome linked recessive disease

50. When a homozygous red flower of snapdragon plant is crossed with a white flowered plant, what colour is produced in F_1?
(a) Red
(b) White
(c) Pink
(d) Patches of Red and white

Exercise-03 Level -03

Assertion & Reason Based Questions

1. **Assertion:** Mendel selected 14 true breeding pea plant varieties.
Reason: A true breeding line is one that have undergone continuous self-pollination and shows stable trait inheritance and expression for several generations.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true

2. **Assertion:** Codominant alleles lack dominant recessive - relationship.
Reason: Codominant alleles show incomplete dominance.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true

3. **Assertion:** Sickle cell anaemia is an example of point mutation.
Reason: In sickle cell anemia, RBCs become sickle shape.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true.

4. **Assertion:** Mendel was successful in his hybridisation experiments.
Reason: Garden pea proved ideal experimental material.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true

5. **Assertion:** Both alleles in codominance are equally - important.
Reason: In codominance the number of phenotypes is the same as the number of alleles.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true

6. **Assertion:** Number of chromosomes in one genome is equal to number of linkage groups.
Reason: Two homologous chromosomes form a linkage group.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true

7. **Assertion:** The genetic makeup of sperm determines the sex of child in humans.
Reason: In humans, males are heterogametic.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true.

8. **Assertion:** Phenylketonuria is a recessive hereditary disease - caused by body's failure to oxidise an amino acid phenylalanine to tyrosine, because of a defective enzyme.
Reason: It results in the presence of phenylalanine in urine.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true.

9. **Assertion:** Human skin colour is controlled by 3 pairs of genes.
Reason: Skin colour in humans show pleiotropic inheritance.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true.

10. **Assertion:** When yellow bodied, white eyed Drosophila females were hybridised with brown-bodied, red eyed males; and F_1 progeny was intercrossed, F_2 ratio deviated from 9:3:3:1.
Reason: When two genes in a dihybrid are on the same chromosome, the proportion of parental gene combinations are much higher than the non-parental type.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true.

11. **Assertion:** The law of independent assortment can be studied by means of dihybrid cross.
Reason: The law of independent assortment is applicable only to linked genes.
(a) Both assertion and reason are true and reason is the correct explanation of assertion.
(b) Both assertion and reason are true but reason is not the correct explanation of assertion.
(c) Assertion is true but reason is false.
(d) Assertion is false but reason is true.

12. **Assertion:** In a monohybrid cross, F_1 generations indicate -dominant characters.
Reason: Dominance occurs only in heterozygous state.
(a) Both (A) and (R) are correct and (R) is the correct explanation of (A).
(b) Both (A) and (R) are correct but (R) is not the correct explanation of (A).
(c) (A) is correct but (R) is not correct.
(d) (A) is not correct but (R) is correct.

13. **Assertion:** Down's syndrome is the trisomy of chromosome number of 21.
Reason: Down syndrome was discovered by Langdon Down in 1866.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

14. **Assertion:** Aneuploidy is the gain or loss of a chromosome in chromosome pool.
Reason: Aneuploidy is failure of segregation of chromatids during cell division cycle.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

15. **Assertion:** Mendelian disorder is alteration or mutation of single gene in gene pool of organism.
Reason: Mendelian disorder sometimes results in form of aneuploidy.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

16. **Assertion:** Point mutation is change in single base pair of DNA.
Reason: Deletion and insertion are reason of abnormalities and aberrations.
(a) If assertion and reason is true and reason is correct explanation of assertion.

(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

17. **Assertion:** In birds, female is one determining the sex of progeny.
Reason: In birds, female show heterogamety.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

18. **Assertion:** Morgan studied the linkage on Drosophila using body colour and eye colour trait.
Reason: Morgan hybridized yellow-bodied female to brown-bodied male.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

19. **Assertion:** Multiple alleles are those having more than two alleles.
Reason: These all alleles represent the same character.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

20. **Assertion:** Law of segregation is dependent on the fact that alleles do not show any blending in cross.
Reason: Characters not visible in F_1 generation are recovered in F_2 generation.
(a) If assertion and reason is true and reason is correct explanation of assertion.
(b) If assertion and reason is true and reason is not correct explanation of assertion.
(c) If assertion is true and reason is false.
(d) If both assertion and reason is false.

Statement Based Questions

21. **Statement I:** Mendel used several true breeding pea lines for experiment.
Statement II: Pure breeding lines are those having undergone continuous cross pollination.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

22. **Statement I:** Mendel conducted artificial pollination in pea plants using contrasting traits.
Statement II: Mendel named progeny as filial generation of parent generation.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

23. **Statement I:** During observation of Mendel's monohybrid cross, he found that only one trait of two plants shows up in F_1 generation.
Statement II: In monohybrid cross, selfing of two tall plants gives 3:1 ratio of progeny.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

24. **Statement I:** Parental allelic pair got separate from each other by the process of mitosis.
Statement II: Segregation of alleles is a random process.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

25. **Statement I:** *Antirrhinum* shows 1:2:1 ratio in F_2 generation.
Statement II: *Antirrhinum* phenotypic ratio differ from genotypic ratio.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

26. **Statement I:** When progeny resembles in-between the two parents, it represents co-dominance.
Statement II: When progeny resembles both the parents, it represents incomplete dominance.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

27. **Statement I:** ABO blood group is classic example of incomplete dominance along with multiple allele.
Statement II: Starch synthesis in peas is example of co-dominance.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

28. **Statement I:** Mendel's laws of inheritance were extended as 'Chromosomal Theory of Inheritance'.
Statement II: Sutton and Boveri both proposed Chromosomal Theory of Inheritance.
(a) Statement I is true and Statement II is false.

(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

29. **Statement I:** Recombination is physical association of genes on a chromosome according to Morgan.
Statement II: In Morgan definition of linkage, linkage is the generation of non-parental gene combination.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

30. **Statement I:** Pleiotropy is expression of single gene into multiple phenotypes.
Statement II: Gene expressing multiple phenotypes is pleiotropic gene.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

31. **Statement I:** If a cell does not go under cytokinesis, then it will cause polyploidy.
Statement II: Aneuploidy is change in subset of chromosome.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

32. **Statement I:** Mutation are associated with inheritance.
Statement II: Mutation cause a number of inheritable disorders.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

33. **Statement I:** Sex linked genes are present on sex-chromosomes.
Statement II: In human, X-chromosome is female chromosome.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

34. **Statement I:** Change in whole set of chromosomes is polyploidy.
Statement II: Polyploidy can be seen in human.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

35. **Statement I:** Turner's Syndrome and Klinefelter's Syndrome are analysed by karyotype of organism.
Statement II: Turner's Syndrome and Klinefelter's Syndrome are syndrome of sex chromosome.
(a) Statement I is true and Statement II is false.
(b) Statement I is false and Statement II is true.
(c) Statement I and Statement II is true.
(d) Statement I and Statement II is false.

Match up Based Questions

36. Match the following:

Column I		Column II	
A	Monohybrid cross	1	9:3:3:1
B	Test cross	2	1:1
C	Dihybrid cross	3	3:1

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

37. Match the following:

Column I		Column II	
A	Henking	1	Named factor
B	de Vries	2	Discovery of X-body
C	Mendel	3	Mutation

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

38. Match the following:

Column I		Column II	
A	ABO blood group	1	Multiple allelism
B	Law of independent assortment	2	Dihybrid cross
C	Law of segregation	3	Monohybrid cross

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

39. Match the following:

Column I		Column II	
A	Multiple allele	1	Many genes governing a single character
B	Pleiotropy	2	More than two allele controlling a single trait
C	Polygenic inheritance	3	Single gene influencing multiple phenotype

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1

(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

40. Match the following w.r.t. Mendelian cross:

Column I		Column II	
A	WW genotype	1	½ heterozygous
B	Ww genotype	2	¼ recessive
C	ww genotype	3	¼ dominant

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

41. Match the following w.r.t. starch synthesis in pea plant:

Column I		Column II	
A	BB gene	1	Intermediate size starch grain
B	bb gene	2	Wrinkled seeds
C	Bb gene	3	High efficiency in starch synthesis

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

42. Match the following:

Column I		Column II	
A	Recombination	1	Very low recombination
B	Tightly linked genes	2	Higher recombination
C	Loosely linked gene	3	Non-parental gene combination

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

43. Match the following w.r.t incomplete dominance:

Column I		Column II	
A	Modified allele	1	No change in enzyme product
B	Unmodified allele	2	Functioning enzyme product
C	Modified equivalent to unmodified allele	3	Non-functioning enzyme product

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

44. Match the following:

Column I		Column II	
A	Alteration in chromosome	1	Pedigree
B	Analysis of trait of generations	2	Chromosomal disorder
C	Abnormal arrangement of chromosome	3	Mendelian disorder
D	Alteration in single gene	4	Mutation

(a) A-1, B-2, C-3, D-4
(b) A-2, B-3, C-4, D-1
(c) A-3, B-4, C-1, D-2
(d) A-4, B-1, C-2, D-3

45. Match the following w.r.t. linkage:

Column I		Column II	
A	Tightly linked gene	1	Male parent
B	White eye character	2	37.2% recombination
C	Red eyed character	3	1.3% recombination
D	Loosely linked gene	4	Female parent

(a) A-1, B-2, C-3, D-4
(b) A-2, B-3, C-4, D-1
(c) A-3, B-4, C-1, D-2
(d) A-4, B-1, C-2, D-3

46. Match the following:

Column I		Column II	
A	1:2:1 phenotype	1	Test cross
B	3:1 phenotype	2	Incomplete dominance
C	1:1 phenotype	3	Monohybrid cross

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

47. Match the following:

Column I		Column II	
A	Non parental gene exchange	1	Recombination
B	Non sister chromatids	2	Polyploidy
C	More than two sets of chromosomes	3	Crossing over

(a) A-1, B-2, C-3
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

48. Match the following:

Column I		Column II	
A	α-Thalassemia	1	Chromosome 21th
B	β-Thalassemia	2	Chromosome 11th
C	Down Syndrome	3	Chromosome 16th

(a) A-1, B-2, C-3,
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

49. Match the following:

Column I (Genetic trait)		Column II (Name of disorders)	
A	Autosomal linked recessive trait	1	Phenylketonuria
B	Sex-linked recessive disease	2	Haemophilia
C	Inborn metabolic error linked to autosomal recessive trait	3	Sickle-cell anemia

(a) A-1, B-2, C-3,
(b) A-2, B-3, C-1
(c) A-3, B-1, C-2
(d) A-3, B-2, C-1

50. Match the following:

Column I		Column II	
A	Polyploidy	1	Human female
B	Additional 21st chromosome	2	Wheat
C	Karyotype of XXY	3	Trisomy
D	Karyotype of XO	4	Human male

(a) A-1, B-2, C-3, D-4
(b) A-2, B-3, C-4, D-1
(c) A-3, B-4, C-1, D-2
(d) A-4, B-1, C-2, D-3

Exercise-04 Previous Year Questions

1. Match List I with List II **(2024)**

List – I		List – II	
A.	Two or more alternative forms of a gene	I.	Back cross
B.	Cross of F_1 progeny with homozygous recessive parent	II.	Ploidy
C.	Cross of F_1 progeny with any of parent	III	Allele
D.	Number of chromosomes sets in plant	IV	Test cross

Choose the correct answer from the options given below:
(a) A – II, B – I, C – III, D – IV
(b) A – III, B – IV, C – I, D – II
(c) A – IV, B – III, C – II, D – I
(d) A – I, B – II, C – III, D – IV

2. In a plant, black seed color (BB/Bb) is dominant over white seed color (bb). In order to find out the genotype of the black seed plant, with which of the following genotype will you cross it? **(2024)**

(a) bb (b) Bb
(c) BB/Bb (d) BB

3. A pink flowered Snapdragon plant was crossed with a red flowered Snapdragon plant. What type of phenotype/s is/are expected in the progeny? **(2024)**

(a) Red Flowered as well as pink flowered plants
(b) Only pink flowered plants
(c) Red, pink as well as white flowered plants
(d) Only red flowered plants

4. Which one of the following can be explained on the basis of Mendel's Law of Dominance? **(2024)**

A. Out of one pair of factors one is dominant and the other is recessive.
B. Alleles do not show any expression and both the characters appear as such in F_2 Generation.
C. Factors occur in pairs in normal diploid plants
D. The discrete unit controlling a particular character is called factor.
E. The expression of only one of the parental characters is found in a monohybrid cross.

Choose the correct answer from the options given below.
(a) A, C, D and E
(b) B, C and D only
(c) A, B, C, D and E
(d) A, B and C only

5. Match List I with List II: **(2024)**

List I		List II	
A.	Down's syndrome	I.	11th chromosome
B.	α-Thalassemia	II.	'X' chromosome
C.	β-Thalassemia	III.	21st chromosome

D.	Klinefelter's syndrome	IV.	16th chromosome

Choose the correct answer from the options given below:

(a) A-II, B-III, C-IV, D-I
(b) A-III, B-IV, C-I, D-II.
(c) A-IV, B-I, C-II, D-III
(d) A-I, B-II, C-III, D-IV

6. The phenomenon of pleiotropism refers to **(2023)**

(a) Presence of two alleles, each of the two genes controlling a single trait.
(b) A single gene affecting multiple phenotypic
(c) More than two genes affecting a single character.
(d) Presence of several alleles of a single Gene controlling a single crossover.

7. Which of the following statements are correct about Klinefelter's Syndrome?

A. This disorder was first described by Langdon (1866).
B. Such an individual has overall Masculine development. However, the Feminine development is also expressed.
C. The affected individual is short statured.
D. Physical, psychomotor and mental development is retarded.
E. Such individuals are sterile.

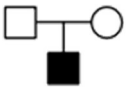
Choose the correct answer from the options given below. **(2023)**

(a) C and D only (b) B and E only
(c) A and E only (d) A and B only

8. Broad palm with single palm crease is visible in a person suffering from **(2023)**

(a) Turner's syndrome
(b) Klinefelter's syndrome
(c) Thalassemia
(d) Down's syndrome

9. Which one of the following symbols represents mating between relatives in human analysis? **(2023)**

(a) 
(b) 
(c) 
(d) 

10. XO type of sex determination can be found in **(2022)**

(a) Drosophila (b) Birds
(c) Grasshoppers (d) Monkey

11. Given below are two statements :

Statement I: Mendel studied seven pairs of contrasting traits in pea plants and proposed the Laws of Inheritance.

Statement II: Seven characters examined by Mendel in his experiment on pea plants were seed shape and colour, flower colour, pod shape and colour, flower position and stem height.

In the light of the above statements, choose the correct answer from the options given below **(2022)**

(a) Both Statement I and Statement II are correct
(b) Both Statement I and Statement II are incorrect
(c) Statement I is correct but Statement II is incorrect
(d) Statement I is incorrect but Statement II is correct

12. Which of the following occurs due to the presence of autosome linked dominant trait? **(2022)**

(a) Sickle cell anaemia
(b) Myotonic dystrophy
(c) Haemophilia
(d) Thalessemia

13. Given below are two statements one is as Assertion (A) and the is labelled as Reason (R).

Assertion (A): Mendel's law of Independent assortment does not hold good for the genes that are located closely on the same chromosome.

Reason (R): Closely located genes assort independently.

In the light of the above statements, choose the correct answer from the options given below **(2022)**

(a) Both (A) and (R) are correct and (R) is the correct explanation of (A)
(b) Both (A) and (R) are correct but (R) is not the correct explanation of (A)
(c) (A) is correct but (R) is not correct
(d) (A) is not correct but (R) is correct

14. The recombination frequency the a & c is 5%, b & c is 15%, b & d is 9%, a & b is 20%, c & d is 24% and a & d is 29%. What will be the sequence of these genes on a linear chromosome? **(2022)**

(a) d, b, a, c (b) a, b, c, d
(c) a, c, b, d (d) a, d, b, c

15. If a colour blind female marries a man whose mother was also colour blind, what are the chances of her progeny having colour blindness? **(2022)**

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

16. The production of gametes by parents, formation of zygote, the f1 and f2 plants can be understood from a diagram called: **(2021)**
(a) Punnett square
(b) Net square
(c) Bullet square
(d) Punch square

17. Identify the wrong statement with reference to the gene 'I' that controls ABO blood groups. **(2020)**
(a) A person will have only two of the three alleles.
(b) When IA and IB are present together, they express same type of sugar.
(c) Allele 'i' does not produce any sugar
(d) The gene (I) has three alleles.

18. Experimental verification of the chromosomal theory of inheritance was done by: **(2020)**
(a) Sutton (b) Boveri
(c) Morgan (d) Mendel

19. Select the correct match **(2020)**

Column - I			Column - II
(a)	Phenylketonuria	-	Autosomal dominant trait
(b)	Sickle cell anaemia	-	Autosomal recessive trait, chromosome-11
(c)	Thalassemia	-	X linked
(d)	Haemophilia	-	Y linked

20. How many true breeding pea plant varieties did Mendel select as pairs, which were similar except in one character with contrasting traits? **(2020)**
(a) 2 (b) 14
(c) 8 (d) 4

21. Chromosomal theory of inheritance was proposed by: **(2020 Covid Re-NEET)**
(a) Bateson and Punnet
(b) T.H. Morgan
(c) Watson and Crick
(d) Sutton and Boveri

22. The number of contrasting characters studied by Mendel for his experiments was: **(2020 Covid Re-NEET)**
(a) 4 (b) 2
(c) 7 (d) 14

23. The best example for pleiotropy is: **(2020 Covid Re-NEET)**
(a) Phenylketonuria
(b) Colour Blindness
(c) ABO Blood group
(d) Skin colour

Answer keys

TOPIC CENTRIC EXERCISE -01 Answer Key

1. (c)	**2.** (c)	**3.** (b)	**4.** (b)	**5.** (b)

TOPIC CENTRIC EXERCISE -02 Answer Key

1. (c)	**2.** (b)	**3.** (c)	**4.** (b)	**5.** (d)

TOPIC CENTRIC EXERCISE -03 Answer Key

1. (a)	**2.** (a)	**3.** (a)	**4.** (c)	**5.** (c)

TOPIC CENTRIC EXERCISE -04 Answer Key

1. (d)	**2.** (a)	**3.** (d)	**4.** (c)	**5.** (b)

TOPIC CENTRIC EXERCISE -05 Answer Key

1. (b)	**2.** (a)	**3.** (d)	**4.** (a)	**5.** (c)

TOPIC CENTRIC EXERCISE -06 Answer Key

1. (c)	**2.** (d)	**3.** (c)	**4.** (c)	**5.** (d)

TOPIC CENTRIC EXERCISE -07 Answer Key

1. (a)	**2.** (c)	**3.** (b)	**4.** (d)	**5.** (b)

Exercise-01 Level -01 Answer Key

1. (b)	**6.** (c)	**11.** (a)	**16.** (d)	**21.** (b)	**26.** (d)	**31.** (d)	**36.** (b)	**41.** (b)	**46.** (d)
2. (c)	**7.** (b)	**12.** (d)	**17.** (d)	**22.** (c)	**27.** (a)	**32.** (b)	**37.** (b)	**42.** (d)	**47.** (b)
3. (c)	**8.** (c)	**13.** (a)	**18.** (b)	**23.** (d)	**28.** (b)	**33.** (a)	**38.** (b)	**43.** (d)	**48.** (b)
4. (d)	**9.** (b)	**14.** (a)	**19.** (b)	**24.** (b)	**29.** (b)	**34.** (a)	**39.** (a)	**44.** (c)	**49.** (d)
5. (b)	**10.** (a)	**15.** (b)	**20.** (a)	**25.** (b)	**30.** (b)	**35.** (d)	**40.** (d)	**45.** (d)	**50.** (c)

Exercise-02 Level -02 Answer Key

1. (d)	**6.** (b)	**11.** (b)	**16.** (d)	**21.** (a)	**26.** (c)	**31.** (a)	**36.** (a)	**41.** (b)	**46.** (a)
2. (a)	**7.** (c)	**12.** (b)	**17.** (d)	**22.** (c)	**27.** (c)	**32.** (a)	**37.** (a)	**42.** (b)	**47.** (a)
3. (c)	**8.** (b)	**13.** (b)	**18.** (b)	**23.** (a)	**28.** (a)	**33.** (a)	**38.** (d)	**43.** (c)	**48.** (c)
4. (c)	**9.** (a)	**14.** (a)	**19.** (d)	**24.** (b)	**29.** (b)	**34.** (a)	**39.** (b)	**44.** (d)	**49.** (d)
5. (c)	**10.** (d)	**15.** (d)	**20.** (d)	**25.** (d)	**30.** (d)	**35.** (a)	**40.** (b)	**45.** (b)	**50.** (c)

Exercise-03 Level -03 Answer Key

1. (a)	**6.** (b)	**11.** (c)	**16.** (b)	**21.** (a)	**26.** (d)	**31.** (c)	**36.** (d)	**41.** (d)	**46.** (b)
2. (c)	**7.** (a)	**12.** (c)	**17.** (a)	**22.** (c)	**27.** (d)	**32.** (c)	**37.** (b)	**42.** (c)	**47.** (c)
3. (b)	**8.** (b)	**13.** (b)	**18.** (b)	**23.** (c)	**28.** (c)	**33.** (c)	**38.** (a)	**43.** (d)	**48.** (d)
4. (b)	**9.** (c)	**14.** (a)	**19.** (b)	**24.** (b)	**29.** (d)	**34.** (a)	**39.** (b)	**44.** (d)	**49.** (d)
5. (c)	**10.** (a)	**15.** (c)	**20.** (b)	**25.** (a)	**30.** (c)	**35.** (c)	**40.** (c)	**45.** (c)	**50.** (b)

Exercise-04 Previous Year Questions

1. (b)	**4.** (a)	**7.** (b)	**10.** (c)	**13.** (c)	**16.** (a)	**19.** (b)	**22.** (c)
2. (a)	**5.** (b)	**8.** (d)	**11.** (a)	**14.** (c)	**17.** (b)	**20.** (b)	**23.** (a)
3. (a)	**6.** (b)	**9.** (a)	**12.** (b)	**15.** (c)	**18.** (c)	**21.** (d)	