



## CHAPTER – 5

# PRINCIPLES OF INHERITANCE AND VARIATION

**Genetics:** the branch of biology that deals with the study of genes, genetic variations and heredity in living organisms.

**Inheritance:** it is the process by which characters are passed on from parent to offsprings and it forms the basis of heredity.

**Variation:** it is the degree by which a progeny differs from their parents.

The cause of variations are:

- o Recombination
- o Reshuffling of genes
- o Mutation

**Mendel's Law of Inheritance:** Mendel conducted seven years of hybridization research on garden pea (*Pisum sativum*) and postulated the rule of inheritance in living beings.

**Selection of Pea plants:** Mendel used garden pea (*Pisum sativum*) for his research for the following reasons:

- o Pea has numerous unique and opposing personalities.
- o The pea plant has a short life cycle.
- o Flowers exhibit self-pollination, with reproductive whorls surrounded by the corolla.
- o It is simple to cross-pollinate pea flowers artificially. The resulting hybrids are fertile.

**His Methodology:** he was successful in his experiments because:

- o He just studied one character at a time.
- o He employed every known means to avoid cross-pollination from unwanted pollen grains.
- o He used mathematics and statistics to analyse the data he acquired.
- o Mendel chose seven opposing garden pea characteristics for his hybridization experiments.
- o Mendel Investigated Contrasting Characters in Pea.

Mendel used true-breeding pea lines to perform artificial hybridization/cross pollination. **True breeding lines** are those that exhibit consistent trait inheritance and undertake continuous self-pollination.

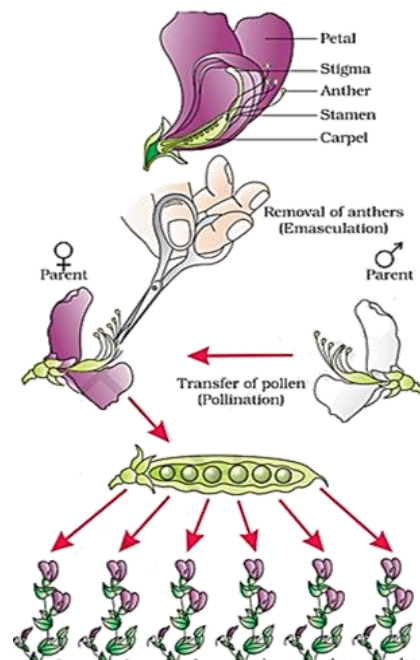
The hybridization experiment comprises emasculation (anther removal) and pollen transfer (pollination).

#### Stages of making a cross in Pea Plant

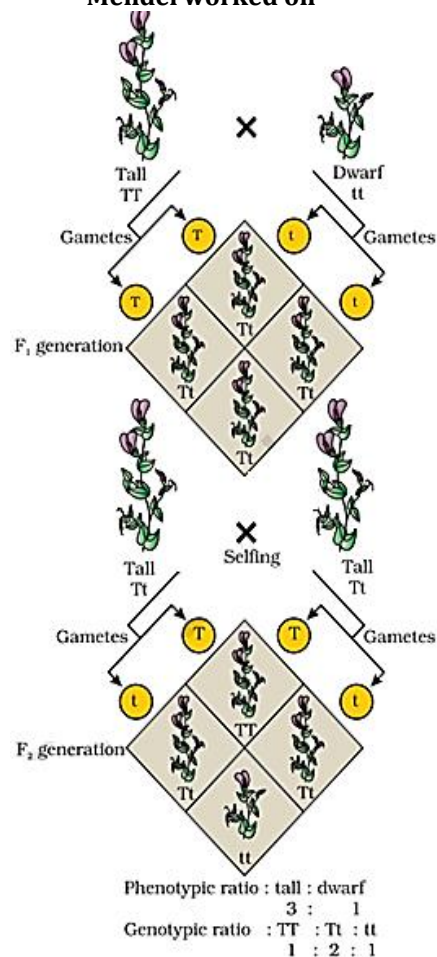
S.No.	Characters	Contrasting Traits
1.	<i>Stem height</i>	<i>Tall/dwarf</i>
2.	<i>Flower colour</i>	<i>Violet/white</i>
3.	<i>Flower position</i>	<i>Axial/terminal</i>
4.	<i>Pod shape</i>	<i>Inflated/constricted</i>
5.	<i>Pod colour</i>	<i>Green/yellow</i>
6.	<i>Seed shape</i>	<i>Round/wrinkled</i>
7.	<i>Seed colour</i>	<i>Yellow/green</i>

#### Monohybrid Cross (Inheritance by one Gene)

- ✓ Mendel crossed tall and dwarf pea plants and gathered all of the seeds.
- ✓ He nurtured all of the seeds to produce plants of the first hybrid generation, known as the F<sub>1</sub> generation.
- ✓ He saw that all of the plants are tall. A similar discovery was made in another pair of features.
- ✓ Mendel self-pollinated the F<sub>1</sub> plants and discovered that some plants in the F<sub>2</sub> generation are similarly small.
- ✓ Dwarf plants account for  $\frac{1}{4}$  of the total, while tall plants account for the  $\frac{3}{4}$  of the total.
- ✓ Mendel called that '**factors**' were responsible for the transfer of gametes from generation to generation. It is now referred to as **genes (unit of inheritance)**.
- ✓ **Alleles are genes that code for a pair of opposing traits.**
- ✓ Each gene is represented by an alphabetical symbol, with a capital letter (TT) for genes expressed in the F<sub>1</sub> generation and a tiny letter (tt) for other genes.
- ✓ Mendel also claimed that in true breeding tall and dwarf varieties, the height allelic pair is homozygous (TT or tt). The genotype is TT, Tt, or tt, represents the phenotypic trait, tall or dwarf.
- ✓ Heterozygous hybrids are those that have alleles that display opposing features (Tt).
- ✓ **The F<sub>2</sub> hybrid's monohybrid ratio is 3:1 (phenotypic) and 1:2:1 (genotypic).**



Seven characters of Pea plant on which Mendel worked on



Representation of monohybrid cross on Punnett Square

**Test Cross:** it is a cross between an individual with dominant trait and a recessive organism in order to know whether the dominant trait is homozygous or heterozygous.

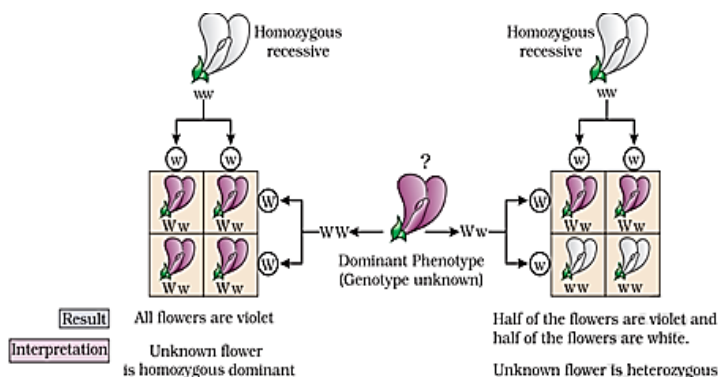


Diagram showing test Cross

## Brush Up Your Understanding

- Q1.** True Breeding lines are those that.
- that exhibit consistent trait inheritance
  - undertake continuous self-pollination.
  - Both (a) and (b)
  - None of the above
- S1. (c)** True breeding lines are those that exhibit consistent trait inheritance and undertake continuous self-pollination.
- Q2.** Methodology of Mendel included.
- He studied one character at a time.
  - He employed every known means to avoid cross-pollination from unwanted pollen grains.
  - He used mathematics and statistics to analyse the data he acquired.
  - All of the above
- S2. (d)** On the basis of observations of the monohybrid cross, Mendel proposed two general rules that are now known as Principles/Laws of Inheritance that are as follows:

**Law of Dominance:** the law states that:

- Characters are controlled by separate units known as factors.
- Factors are always found in pairs.
- In a dissimilar pair of factors, one dominates the other.

**Dominance:** Dominance occurs when a factor (allele) manifests itself in the presence or absence of its dominant factor. It generates a fully functioning enzyme that expresses it precisely.

**Recessive:** It can only manifest in the absence of or recessive factor allele. When present with its dominant allele, i.e., in a heterozygous state, it generates an incomplete deficient enzyme that fails to express itself.

**Law of segregation:** the law states that:

- Alleles do not mix, and both characters are recovered after gamete creation, as in the F<sub>2</sub> generation.
- Traits segregate (separate) from one another during gamete formation and transmit to various gametes.

- Homozygous individuals generate identical types of gametes, whereas heterozygous individuals create gametes with distinct characteristics.

**Incomplete Dominance:** It is a post-Mendelian finding, incomplete dominance occurs when none of the two alleles is dominant, resulting in expression in the hybrid that is a fine mixture or intermediate between the expressions of the two alleles.

There are two varieties of pure breeding plants in snapdragon (*Mirabilis jalapa*), red flowered and white flowered. Pink flowers are produced by crossing the two. When selfed, the F<sub>2</sub> generation has one red, two pinks, and one white. The pink flowers is the result of incomplete dominance.

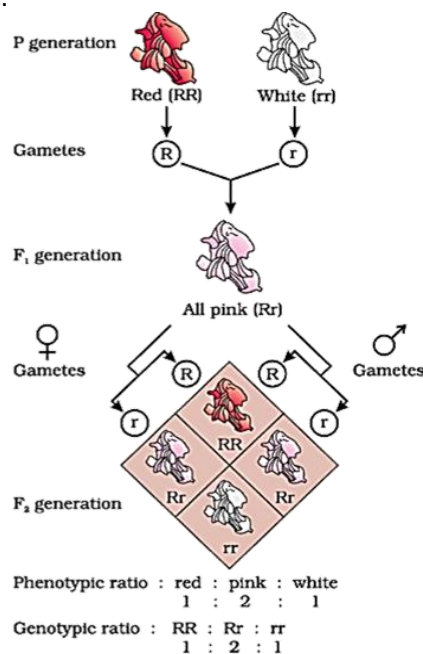


Diagram showing incomplete dominance in snapdragon



**Co-dominance:** It is caused by two alleles that do not have a dominant-recessive connection and both express themselves in the organism.

- ✓ ABO blood grouping in humans is governed by gene  $I$ . There are three alleles in the gene:  $I^A$ ,  $I^B$ , and  $i$ . Any two of the three alleles  $I^A$ ,  $I^B$  are dominant over  $i$  in any individual.
- ✓ The sugar polymers that protrude from the surface of the plasma membrane of red blood cells are regulated by the gene.
- ✓ Because of co-dominance, when  $I^A$  and  $I^B$  are present together, they both express their own forms of sugars.

**Multiple alleles:** They are several variants of a Mendelian factor or gene that occur on the same gene locus and are dispersed in the gene pool in various animals, with an organism having only two alleles and a gamete bearing just one allele. ABO blood categorization is another example of multiple alleles.

## Brush Up Your Understanding

- Q1.** Mendelian ratio 9:3:3:1 is due to.  
 (a) Law of segregation  
 (b) Law of purity of gametes  
 (c) Law of independent assortment  
 (d) Law of unit character
- S1. (c)**
- Q2.** In case of co-dominance the monohybrid ratio of phenotypes in  $F_2$  generation is.  
 (a) 3 : 1  
 (b) 1 : 2 : 1  
 (c) 1 : 1 : 1 : 1  
 (d) 2 : 2
- S2. (b)**

### Inheritance of two genes (Dihybrid cross)

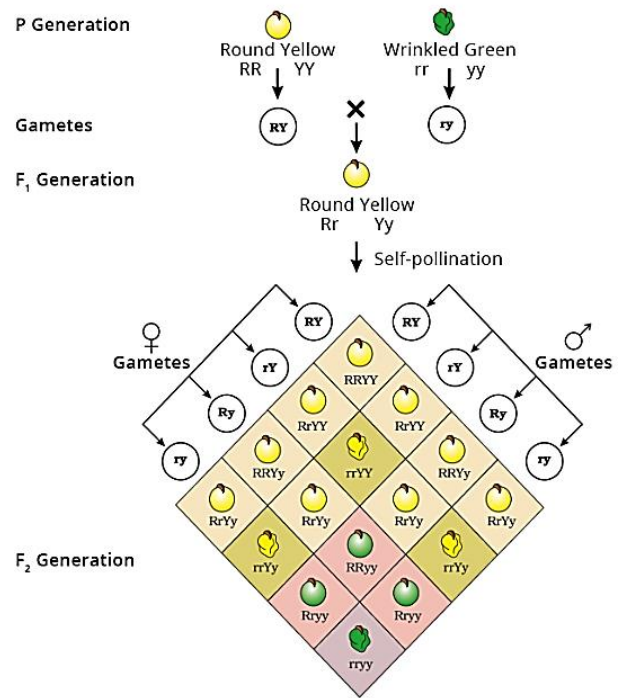
When two pairs of characters are taken into account is a dihybrid cross.

**Law of Independent Assortment:** the law states that. When two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of segregation of the other pair of characters."

Due to the separate assortment of characteristics for seed shape (round, wrinkled) and seed colour (yellow and green) in Dihybrid crosses, two novel combinations, round green & wrinkled yellow, are produced.

The 9:3:3:1 ratio may be calculated by combining 3 yellow: 1 green and 3 round: 1 wrinkled. This is how the derivation is written: (1 Wrinkled: 3 Round) (3 yellow: 1 green) = 9 round, yellow: 3 wrinkled, yellow: 3 round, green: 1 wrinkled, green: 1 wrinkled, green: 1 wrinkled, green: 1

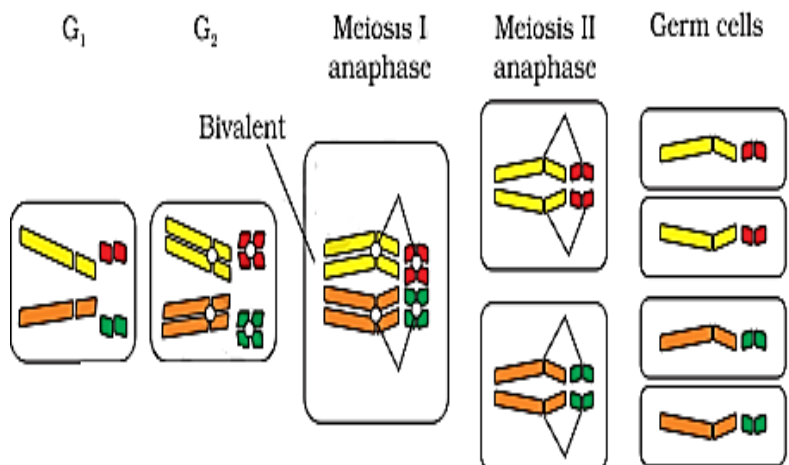
### Mendel's Dihybrid Cross



Results of a dihybrid cross

### Chromosomal Theory of Inheritance

- ✓ Both chromosomes and genes exist in pairs. A gene pair's two alleles are found on the same locus on homologous chromosomes.
- ✓ Sutton and Boveri contended that the pairing and separation of two chromosomes would result in the segregation of a pair of factors (genes) they carried.
- ✓ Sutton coined the term "chromosomal theory of inheritance" after combining knowledge of chromosomal segregation with Mendelian concepts.



Segregation of chromosomes during gamete formation

## Linkage and Recombination

When two genes in a Dihybrid cross were located on the same chromosome, the fraction of parental gene combination was much greater than the non-parental kind.

Morgan described these to the two genes' physical relationship or **linkage**, and developed the term linkage to characterise the physical association of genes on the same chromosome.

**Recombination** is the process through which non-parental gene combinations are generated during a Dihybrid cross.

When genes are found on the same chromosome, they are closely connected and exhibit relatively little recombination.

## Brush Up Your Knowledge

- Q1. The term linkage was coined by.  
(a) Correns  
(b) Tschermak  
(c) Morgan  
(d) All of the above
- S1. (c).
- Q2. The phenotypic ratio of the  $F_2$  generation of the Mendel's dihybrid cross was.  
(a) 1:2:1 (b) 2:1  
(c) 9:3:3:1 (d) 1:1
- S2. (c)

## Sex Determination

In 1891, Henking discovered a trace of a unique nuclear structure in a few insects. He also discovered that this unique nuclear structure is only seen in 50% of sperms. He referred to this as an **X body**. He couldn't understand its relevance.

Later, it was discovered that the ovum that receives sperms with an X body becomes female, while those that do not receive an X body become males, hence this X body was called the sex chromosome, and the other chromosomes were called autosomes.

**Humans and other species exhibit XY sex determination, whereas some insects, such as Drosophila, exhibit XO sex determination.**

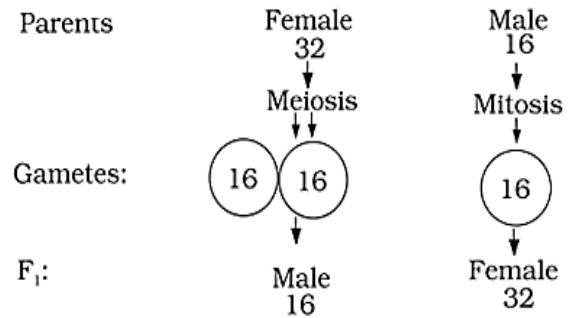
Males produce two types of gametes in both methods of sex determination, either with or without an X chromosome, or some with an X chromosome and some with Y chromosomes. This is called **male heterogamety**.

In birds, the ZW form of sex determination is present; females produce two different types of gametes in terms of sex chromosomes; this type of sex determination is known as **female heterogamety**.

## Sex Determination in Honeybee

Sex determination in Honey Bee is based on the number of set 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 unfertilized egg develops as a male (drone) by means of parthenogenesis, this means that the

males will have half the number of chromosomes than that of female, the females have 32 chromosomes and males are haploid having 16 chromosomes, this is called **haploid sex determination**.



Sex determination in honey bee

## Sex Determination in Human Beings

Humans show XY type of sex determination. Autosomes are 22 pairs of chromosomes that are identical in male and female. Females have two X chromosomes, whereas males have XY chromosomes. During spermatogenesis, males produce two types of gametes (sperms), with half carrying the Y chromosome and the other half carrying the X chromosome. Females produce only one type of gamete (ovum) with X chromosomes.

When Y chromosome sperm fertilises an egg, the child is male; when X chromosome sperm fertilises an egg, the child is female.

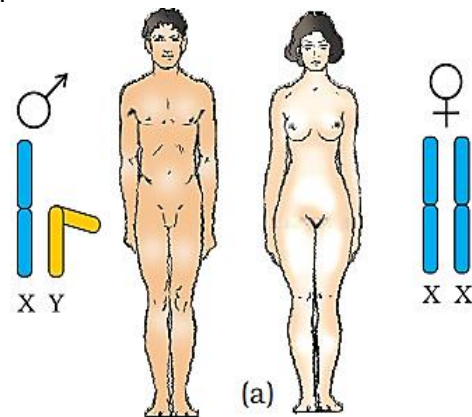


Diagram showing male and female chromosome and they look like

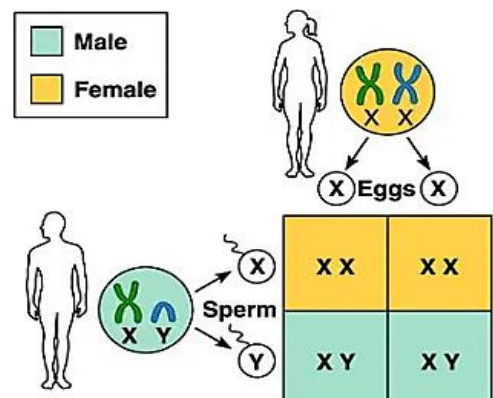


Diagram showing XY sex determination in humans

## Brush Up Your Understanding

- Q1.** In honey bee, an unfertilised egg develops into a.  
 (a) Worker (b) Male  
 (c) Female (d) All of the above
- S1. (b)**
- Q2.** In humans, which among the following produces a heterogamete?  
 (a) Male  
 (b) Female  
 (c) Both (a) and (b)  
 (d) None of the above

**S2. (a)**

- **Mendelian disorders:** These are the result of a single gene change. They are passed down via generations using Mendelian inheritance principles. E.g
- o **Haemophilia:** A sex-linked recessive condition in which a tiny injury causes non-stop bleeding in an affected individual. Females who are heterozygous (carriers) for the condition can pass it on to their son. The likelihood of a female becoming a haemophilic is exceedingly unlikely since the mother must be at least a carrier and the father must be a haemophilic (unviable in the later stage of life).
- **Sickle cell anaemia:** it is an autosomal recessive condition in which mutant haemoglobin molecules polymerize under low oxygen tension, causing the form of the RBC to shift from a biconvex disc to an elongated sickle-like structure. The deficiency is produced by the replacement of Glutamic acid (Glu) for Valine (Val) at the sixth position of the haemoglobin molecule's beta-globin chain. The amino acid change in the globin protein is caused by a single base substitution from GAG to GUG at the sixth codon of the beta globin gene.
- o **Phenylketonuria:** Inborn error of metabolism inherited as an autosomal recessive trait. The afflicted person is deficient in an enzyme that transforms the amino acids phenylalanine to tyrosine. As a result, phenylalanine accumulates and is transformed into phenyl pyruvic acid and other derivatives, which causes mental retardation.
- o **Thalassemia:** it is an autosome linked recessive blood disease that is transmitted to parents to offspring when both the partners are an unaffected carrier for the gene (or heterozygous). The defect is either due to mutation or deletion that results in the reduced rate of

**Mutation:** is a phenomenon that causes the DNA sequence of an organism to change, resulting in a change in genotype and phenotype.

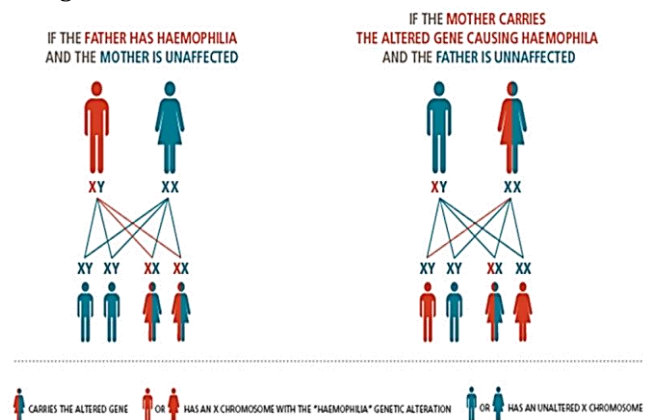
**Point mutations** are those that occur as a result of a change in a single base pair of DNA, such as Sickle cell anaemia.

**Pedigree Analysis:** Pedigree analysis refers to the study of traits in several generations of a family. A family tree depicts the inheritance of a certain trait over numerous generations. It is used to trace the ancestry of a certain trait, aberration, or illness.

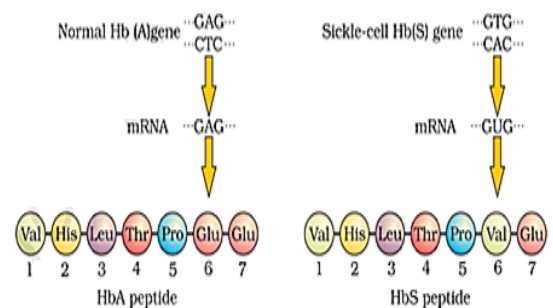
**Genetic Disorders:** They are transmitted as the affected individual is sterile.

This is always dominant in nature.

Broadly, genetic disorders may be grouped into two categories –



**Diagram showing haemophilic pattern and its inheritance**



**Diagram representing sickle cell anaemia**

synthesis of one of the globin chains (alpha or beta) that make up haemoglobin. The cause of the formation of abnormal haemoglobin molecules resulting into anaemia is a characteristic of the disease.

## Brush Up Your Knowledge

- Q1.** The cause of colourblindness is due to.  
 (a) Mutation in genes present on Y-chromosome  
 (b) Mutation in genes present in X-chromosome  
 (c) Both (a) and (b)  
 (d) None of the above
- S1. (b)**

- Q2.** Which of the following is correct about thalassemia?
- (a) It is an autosome linked recessive blood disease
  - (b) Both the partners are unaffected carrier for the gene
  - (c) The defect is either due to mutation or deletion
  - (d) All of the above

**S2. (d)**

**Chromosomal disorders:** These are caused by the absence or presence of one or more chromosomes, or by an aberrant arrangement of one or more chromosomes. In nature, they might be recessive or dominant.

**Aneuploidy** occurs when chromatids fail to segregate during cell division, resulting in chromosomal loss or gain.

The failure of cytokinesis results in two sets of chromosomes, known as **polyploidy**.

- o **Down's Syndrome:** it is due to the existence of an extra copy of chromosome 21. The afflicted person is short and stocky, with a tiny rounded head, wrinkled tongue, and half parted mouth. Mental growth is slowed.
- o **Klinefelter's syndrome:** because of the existence of an extra copy of the X-chromosome (XXY). Such people have overall male growth, but they also exhibit feminine

development (breast development, i.e., Gynaecomastia). They are sterile.

- o **Turner's syndrome:** caused by the lack of one of the X chromosomes 45 with XO, are infertile because their ovaries are underdeveloped. They don't have any secondary sexual characteristics.

### Brush Up Your Knowledge

- Q1.** The disease that is caused due to a gain of an extra copy of chromosome 21 is.

- (a) Turner's syndrome
- (b) Klinefelter's syndrome
- (c) Down's syndrome
- (d) All of the above

**S1. (c)**

- Q2.** In which of the following chromosomal disorder, the individual is devoid of an X-chromosome?

- (a) Klinefelter's syndrome
- (b) Down's syndrome
- (c) Turner's syndrome
- (d) None of the above

**S2. (c)**



## SUMMARY

Genetics is the study of principles and mechanism of heredity and variation. Gregor Johann Mendel is known as 'father of Genetics'. Inheritance is the process by which characters are passed on from parent to progeny. It is the basis of heredity. Variation is the degree by which progeny differ from their parents. Variation may be in terms of morphology, physiology, cytology and behaviouristic traits of individual belonging to same species.

Mendel conducted hybridization experiments on garden pea (*Pisum sativum*) for seven years and proposed the law of inheritance in living organisms. In monohybrid cross, Mendel crossed tall and dwarf pea plant and collected all the seeds obtained from this cross. He grew all the seeds to generate plants of first hybrid generation called  $F_1$  generation. He observed that all the plants are tall. Similar observation was also found in other pair of traits. Based on observations of monohybrid cross, Mendel proposed two laws of inheritance, namely **Law of dominance and law of segregation**.

**Test cross** is the cross between an individual with dominant trait and a recessive organism in order to know whether the dominant trait is homozygous or heterozygous.

Incomplete dominance is a post Mendelian discovery. It is the phenomenon of neither of the two alleles being dominant so that expression in the hybrid is a fine mixture or intermediate between the expressions of two alleles. E.g snapdragon

**Co-dominance** is the phenomenon of two alleles lacking dominance-recessive relationship and both expressing themselves in the organism. E.g ABO blood group in humans.

Multiple alleles are multiple forms of a Mendelian factor or gene which occur on the same gene locus distributed in different organisms in the gene pool with an organism carrying only two alleles and a gamete only one allele. ABO blood grouping also provides a good example of multiple alleles.

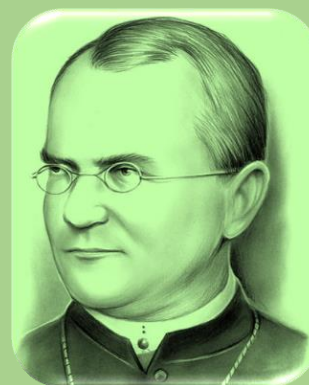
On the basis of a dihybrid cross, Mendel proposed the Law of **Independent Assortment**.

Chromosome theory of inheritance was first proposed by **W.S. Sutton & Boveri in 1902**.

Linkage is the phenomenon of certain genes staying together during inheritance through generations without any change or separation. This is due to their location on the same chromosomes. Linkage maps, therefore, corresponded to arrangement of genes on a chromosome. The recombination frequency of the test cross progeny is always lower than 50%. Therefore, if any two genes are completely linked, their recombination frequency is almost 0%.

Many genes were linked to sexes also, and called as sex-linked genes. The two sexes (male and female) were found to have a set of chromosomes which were common, and another set which was different. The chromosomes which were different in two sexes were named as sex chromosomes. The remaining set was named as autosomes. In humans, a normal female has 22 pairs of autosomes and a pair of sex chromosomes (XX). A male has 22 pairs of autosomes and a pair of sex chromosome as XY. In chicken, sex chromosomes in male are ZZ, and in females are ZW.

A change in the genetic material is called mutation. A point mutation is a change of a single base pair in DNA. Inheritable mutations can be studied by generating a pedigree of a family. Polyploidy is the change in whole set of chromosomes while aneuploidy is the change in a subset of chromosome number.



## IMPORTANT POINTERS

### Mendel is called as Father of Genetics

Genetics is the study of genes, genetic code heredity, and variations.

The phenotypic ratio in Mendel's monohybrid cross was 3:1 and the genotypic ratio was 1:2:1.

In a typical test cross an organism showing a dominant phenotype (and whose genotype is to be determined) is crossed with the recessive parent instead of self-crossing.

There are two exceptions of law of dominance, incomplete dominance and co-dominance.

In incomplete dominance (Snapdragon), the phenotypic and the genotypic ratio are same and that is 1:2:1.

ABO blood grouping in humans is an example of Multiple allelism.

Starch synthesis in pea seeds is controlled by one gene (it has two alleles).

The phenotypic ratio in Mendel's dihybrid cross is 9:3:3:1.

The phenomenon of linkage was studied by the scientist T.H. Morgan using the common fruit fly or *Drosophila melanogaster*.

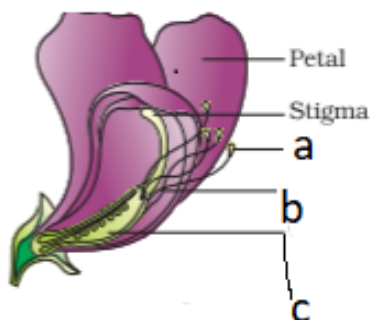
Colour blindness and haemophilia are X-linked diseases,

Trisomy of 21st chromosome causes Down's syndrome while in Turner's syndrome one of the X-chromosomes is missing (XO), XXY is the condition seen in Klinefelter's syndrome.

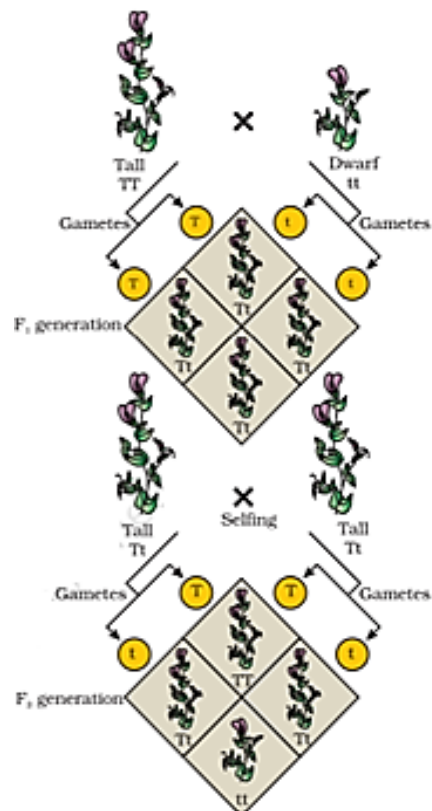


## MULTIPLE CHOICE QUESTIONS

- Q1.** Which of the following subjects deals with the inheritance as well as the variation of characters from parents to offspring?  
 (a) Biotechnology  
 (b) Genetics  
 (c) Molecular biology  
 (d) All of the above
- Q2.** Mendel conducted his hybridization experiments on which of the following plant?  
 (a) Tomato  
 (b) Potato  
 (c) Pea  
 (d) All of the above
- Q3.** Which of the following contrasting characters were used by Mendel for his hybridisation studies?  
 (a) Stem height  
 (b) Flower colour  
 (c) Pod colour  
 (d) All of the above
- Q4.** What was the ratio of the tall and short plants in the F<sub>2</sub> generation in the experiments performed by Mendel on tall and short pea plants?  
 (a) 1:1  
 (b) 2:1  
 (c) 3:1  
 (d) 4:1
- Q5.** What is the meaning of emasculation?  
 (a) It is the removal of pistil from a flower  
 (b) It is the removal of anther from a flower  
 (c) It is the removal of petals from a flower  
 (d) It is the removal of sepals from a flower
- Q6.** Given below is a diagram of a flower, name part a, b and c.



- (a) a. Carpel, b. stamen, c. anther  
 (b) a. Anther, b. stamen, c. carpel  
 (c) a. Stamen, b. carpel, c. anther  
 (d) a. Carpel, b. stamen, c. anther
- Q7.** In a cross between a tall pea plant and dwarf pea plant, what was the ratio that Mendel obtained in the F<sub>2</sub> generation?  
 (a) 1:1  
 (b) 2:1  
 (c) 3:1  
 (d) 4:1
- Q8.** Look at the diagram of a Mendel's monohybrid cross. How many plants in the F<sub>2</sub> generation are homozygous tall?



- (a) 1  
 (b) 2  
 (c) 3  
 (d) 4
- Q9.** In the diagram given in question number 8, state the genotypic ratio of tall and dwarf plants in the F<sub>2</sub> generation.  
 (a) 1: 3:1  
 (b) 1:2:1  
 (c) 3:2:1  
 (d) 2:3:1
- Q10.** In a test cross an organism with a dominant phenotype and whose genotype is to be determined is crossed with a.  
 (a) Recessive parent  
 (b) Dominant parent  
 (c) Self-crossed  
 (d) Cross-pollinated
- Q11.** Mendel's Law of Dominance states that.  
 (a) Characters are controlled by factors  
 (b) Factors occur in pairs  
 (c) In dissimilar pair of factors, one is dominant while the other is recessive  
 (d) All of the above
- Q12.** Which of the following shows incomplete dominance?  
 (a) Pea  
 (b) Snapdragon  
 (c) Antirrhinum  
 (d) Both (b) and (c)
- Q13.** What is the genotypic ratio when a true-breeding red flower plant is crossed with a true-breeding white flower plant showing incomplete dominance?  
 (a) 3:1  
 (b) 1:2:1  
 (c) 2:1  
 (d) 4:1

- Q14.** Which of the following is an example of incomplete dominance in humans?  
 (a) Presence of curly hair  
 (b) Tay-Sachs disease  
 (c) Human skin colour  
 (d) All of the above
- Q15.** Which of the following gene is responsible for ABO blood group in humans?  
 (a) Gene *Y* (b) Gene *X*  
 (c) Gene *I* (d) Gene *O*
- Q16.** Which of the following I governing ABO blood group in humans show complete dominance?  
 (a)  $I^A$  (b)  $I^B$   
 (c) *I* (d) Both (a) and (b)
- Q17.** What was the phenotypic ratio of Mendel's dihybrid cross?  
 (a) 1:2:2:1 (b) 3:2:2:1  
 (c) 9:3:3:1 (d) 1:3:3:9
- Q18.** Which of the following diseases is an example pleiotropy?  
 (a) Sickle cell anaemia  
 (b) Downs syndrome  
 (c) Phenylketonuria  
 (d) Klinefelters syndrome
- Q19.** What are the phenotypic effects seen in a person suffering from the disease phenylketonuria?  
 (a) Mental retardation  
 (b) Reduction in hair  
 (c) Reduction in skin pigmentation  
 (d) All of the above
- Q20.** The specific nuclear structure observed by Henking in spermatogenesis in insects was.  
 (a) Y-body (b) X-body  
 (c) A-body (d) B-body
- Q21.** Which type of sex-determination is observed in grasshopper?  
 (a) XX-type (b) XO-type  
 (c) XY-type (d) YY-type
- Q22.** ZW chromosomes are present in.  
 (a) Female birds  
 (b) Male birds  
 (c) Female grasshopper  
 (d) Male grasshopper
- Q23.** What is the number of chromosomes in a human female?  
 (a) 22 + XX (b) 22 + XY  
 (c) 22 + XO (d) 22 + XXY
- Q24.** Which of the following is correct about sex determination in honey bee?  
 (a) The union of sperm and an egg develops into a queen  
 (b) The union of sperm and an egg develops into a worker  
 (c) Both (a) and (b)  
 (d) None of the above
- Q25.** An unfertilised egg in a honey bee develops into a.  
 (a) Male honey bee  
 (b) Drone  
 (c) The development is through parthenogenesis  
 (d) All of the above
- Q26.** The number of chromosomes in male and female honeybee is.  
 (a) 32, 16 (b) 16, 16  
 (c) 16, 32 (d) 32, 32
- Q27.** Which of the following are the common causes of mutations?  
 (a) Smoking (b) Sunlight  
 (c) Radiation (d) All of the above
- Q28.** Which of the following type of mutations is transmitted to descendant's cells by DNA replication?  
 (a) Germinal mutations  
 (b) Somatic mutation  
 (c) Both (a) and (b)  
 (d) None of the above
- Q29.** Which of the following is Mendelian disorder?  
 (a) Cystic fibrosis (b) Sickle cell anaemia  
 (c) Haemophilia (d) All of the above
- Q30.** Which of the following is a sex-linked recessive disorder?  
 (a) Down's syndrome (b) Colour blindness  
 (c) Klinefelter's syndrome (d) Turner's syndrome
- Q31.** Which of the following provides a strong tool to trace the inheritance of a specific trait, abnormality or a disease in human genetics?  
 (a) Mutation study  
 (b) Pedigree study  
 (c) Sex determination study  
 (d) None of the above
- Q32.** Mendelian disorders can be.  
 (a) Dominant (b) Recessive  
 (c) Both (a) and (b) (d) None of the above
- Q33.** Colour-blindness is caused due to mutation in the.  
 (a) Y-chromosome (b) X-chromosome  
 (c) XY-chromosome (d) None of the above
- Q34.** Who among the following are more effected by colour-blindness?  
 (a) Males (b) Females  
 (c) Both (a) and (b) (d) None of the above
- Q35.** A woman is the carrier of colour-blindness. What are the chances of her son being colour-blind?  
 (a) 100% (b) 50%  
 (c) 25% (d) 10%

- Q36.** Which of the following can transmit the disease haemophilia to her offsprings?  
 (a) Homozygous female (b) Homozygous male  
 (c) Heterozygous female (d) Heterozygous male
- Q37.** Which of the following is an autosomal linked recessive trait?  
 (a) Haemophilia (b) Colour-blindness  
 (c) Sickle cell anaemia (d) All of the above
- Q38.** In Sickle Cell anaemia which of the following will show a diseased phenotype?  
 (a)  $Hb^A Hb^A$  (b)  $Hb^A Hb^S$   
 (c)  $Hb^S Hb^S$  (d) All of the above
- Q39.** In sickle cell anaemia, which of the following amino acid gets substituted by valine at the sixth position of the beta globin chain of haemoglobin molecule?  
 (a) Methionine (b) Glutamic acid  
 (c) Lysine (d) Aspartic acid
- Q40.** In sickle cell anaemia, the RBC change their shape from.  
 (a) Biconvex disc to elongated sickle like structure  
 (b) Biconcave disc to rectangular sickle like structure  
 (c) Biconcave disc to elongated sickle like structure  
 (d) Biconvex disc to square sickle like structure
- Q41.** Which of the following disease is an inborn error of metabolism and is also inherited as an autosomal recessive trait?  
 (a) Thalassemia (b) Sickle cell anaemia  
 (c) Phenylketonuria (d) All of the above
- Q42.** Beta Thalassemia is controlled by.  
 (a) HBA1 gene on chromosome 16  
 (b) HBA2 gene on chromosome 16  
 (c) HBB gene on chromosome 11  
 (d) All of the above
- Q43.** Down's syndrome is caused due to.  
 (a) Loss of chromosome 21  
 (b) Gain of extra copy of chromosome 21  
 (c) Extra copy of a X-chromosome  
 (d) Extra copy of a Y-chromosome
- Q44.** Which of the following disease is caused due to absence of one of the X-chromosome?  
 (a) Klinefelter's syndrome (b) Down's syndrome  
 (c) Turner's syndrome (d) None of the above
- Q45.** Physical appearance of characters is called.  
 (a) Genotype (b) Phenotype  
 (c) Both (a) and (b) (d) None of the above
- Q46.** What symptoms does a person affected with Down's syndrome shows?  
 (a) Furrowed tongue  
 (b) Impairment of psychomotor skills

- (c) Partially opened mouth  
 (d) All of the above

- Q47.** Which of the following is correct about linkage?  
 (a) It results in the formation of recessive phenotype  
 (b) It results in the formation of dominant phenotype  
 (c) It results in the formation of parental phenotype  
 (d) All of the above
- Q48.** The term linkage was coined by.  
 (a) Darwin  
 (b) Haeckel  
 (c) Morgan  
 (d) Lamarck
- Q49.** In which of the following phases of cell division, crossing over occurs?  
 (a) Prophase I  
 (b) Metaphase I  
 (c) Prophase II  
 (d) Metaphase II
- Q50.** Which of the following law was proposed by Mendel?  
 (a) Law of dominance  
 (b) Law of segregation  
 (c) Law of independent assortment  
 (d) All of the above

### ASSERTION AND REASONING

**Direction:** in the following questions, a statement of assertion (A) is followed by a statement of reason (R). Choose the correct option among a, b, c and d.

- Q1. Assertion (A):** there are three pairs of alleles, which responsible to control the human skin colour.  
**Reason (R):** The inheritance of human skin colour called as Polygenic Inheritance.  
 (a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
 (b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
 (c) Assertion (A) is true but reason(R) is false  
 (d) Assertion (A) is false but reason(R) is true
- Q2. Assertion (A):** In human female , XX is the sex chromosomal configuration.  
**Reason (R):** The determination of the sex is done by both the parents.  
 (a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
 (b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
 (c) Assertion (A) is true but reason(R) is false  
 (d) Assertion (A) is false but reason(R) is true

- Q3. Assertion (A):** The Down's syndrome is a Mendelian disorder.  
**Reason (R):** Down's Syndrome disease is caused due to trisomy of chromosome number 21.
- (a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
 (b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
 (c) Assertion (A) is true but reason(R) is false  
 (d) Assertion (A) is false but reason(R) is true
- Q4. Assertion (A):** Thalassaemia is an autosome-linked recessive blood disease.  
**Reason (R):** The defect could be due to either mutation or deletion which ultimately results in reduced rate of synthesis of one of the globin chains ( $\alpha$  and  $\beta$  chains) that make up haemoglobin
- (a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)

- (b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
 (c) Assertion (A) is true but reason(R) is false  
 (d) Assertion (A) is false but reason(R) is true

### TRUE AND FALSE

- Q1.** Morgan worked with the tiny fruit flies, *Drosophila melanogaster*, which were found very suitable for such studies. They could be grown on simple synthetic medium in the laboratory.
- Q2.** Human Skin colour is classical example of pleiotropy.
- Q3.** Cystic fibrosis and sickle cell anaemia are Mendelian disorders.
- Q4.** Haemophilia is an inborn error of metabolism.

## PRACTICE QUESTIONS

- Q1.** Variations are.  
 (a) Degree by which progeny differs from their parents  
 (b) Degree by which progeny similar to their parents  
 (c) Process by which characters are passed on from parent to progeny  
 (d) True breeding lines
- Q2.** Mendel found that the F1 always resembled either one of the parents and that the trait of the other parent was not seen in them. This is due to.  
 (a) Segregation (b) Dominance  
 (c) Partial dominance (d) Unit factor
- Q3.** In monohybrid cross the allele do not show any blending and that both the characters are recovered as such in F2 generation. This statement is explained on the basis of.  
 (a) Dominance  
 (b) Segregation  
 (c) Independent assortment  
 (d) All the above
- Q4.** In monohybrid cross proportion of 3 : 1 explains:  
 (a) Dominance (b) Segregation  
 (c) Both (a) and (b) (d) Unit factor
- Q5.** Theoretically, the modified allele could be responsible for the production of.  
 (a) less efficient enzyme  
 (b) A non functional enzyme  
 (c) Non enzyme at all  
 (d) All the above
- Q6.** The modified allele is equivalent to the unmodified allele when it produces.  
 (a) Normal enzyme  
 (b) A non functional enzyme  
 (c) No enzyme at all  
 (d) Inactive enzyme
- Q7.** Recessive traits are seen due to.  
 (a) Formation of non-functional enzyme  
 (b) Enzyme is not produced  
 (c) Both (a) and (b)  
 (d) Formation of functional enzyme
- Q8.** A diploid organism is heterozygous for 4 loci, how many types of gametes can be produced?  
 (a) 8 (b) 16  
 (c) 2 (d) 32
- Q9.** When a cross is made between tall plant with yellow seed (TtYy) and tall plant with green seed (Tt yy), what proportion of phenotype in the offspring could be expected to be tall and green.  
 (a) 25% (b) 12.5%  
 (c) 37.5% (d) 50%
- Q10.** Which of the following was/were applied first time to problems in biology during Mendel's investigations into inheritance?  
 (a) Statistical analysis  
 (b) Mathematical logic  
 (c) Computational devices  
 (d) Both (a) and (b)



- Q11.** In a large number of insects, the mechanism of sex determination is of.  
 (a) XO type (b) XY type  
 (c) ZW type (d) All the above
- Q12.** Male heterogamety found in.  
 (a) Human (b) Grasshopper  
 (c) Many birds (d) Both (a) and (b)
- Q13.** Who among the following united the knowledge of chromosomal segregation with Mendelian principles and called it chromosomal theory of inheritance  
 (a) Bateson (b) Boveri  
 (c) Sutton (d) Correns
- Q14.** Which of the following structure was discovered by Henking?  
 (a) y-body (b) Bar body  
 (c) x-body (d) Nu-body
- Q15.** In some insects half of the sperms possess X chromosome along with autosomes while half of the sperms carries.  
 (a) Only autosomes  
 (b) y chromosome along with autosome  
 (c) x chromosome only  
 (d) x chromosome along with autosomes
- Q16.** Female heterogamety can be seen in  
 (a) Human beings (b) Drosophila  
 (c) Hen (d) Honey bees
- Q17.** Study of family history about inheritance of a particular trait in several generations of a family called.  
 (a) Phylogeny  
 (b) Ontogeny  
 (c) Pedigree analysis  
 (d) Cladistics
- Q18.** Genetic disorders determined by alteration or mutation in single gene are known as  
 (a) Chromosomal disorders  
 (b) Mendelian disorders  
 (c) Non inheritable disorders  
 (d) All above
- Q19.** Which of the following is not a Mendelian disorder?  
 (a) Haemophilia (b) Colour blindness  
 (c) Down's syndrome (d) Sickle cell anaemia
- Q20.** Chromosomal disorders arise due to.  
 (a) Absence of one or more chromosomes  
 (b) Excess of one or more chromosomes  
 (c) Abnormal arrangement of one or more chromosomes  
 (d) All the above
- Q21.** Select the incorrect statement for Gregor Mendel.  
 (a) He conducted hybridization experiments on garden pea for seven years.  
 (b) He applied statistical analysis and mathematical logic for the first time to the problems in biology.  
 (c) His experiments had a small sampling size.  
 (d) He conducted artificial cross-pollination experiments using several truebreeding pea lines.
- Q22.** Based on observation on monohybrid crosses Mendel draw some conclusion. Which of the following is not correct?  
 (a) Characters are controlled by discrete units called factors  
 (b) Factors occur in pairs  
 (c) In a similar pair of factors one member of the pair dominates the other  
 (d) The postulate of dominance also explains the proportion of 3 : 1 obtained at the F<sub>2</sub>
- Q23.** Which of the following is not concerned with sickle cell anaemia?  
 (a) Sixth position of  $\beta$ -chain  
 (b)  $\alpha$  chain of Hb  
 (c) Valine  
 (d) Haemoglobin
- Q24.** Incomplete dominance can be seen in.  
 (a) *Antirrhinum*  
 (b) *Pisum sativum*  
 (c) Both (a) and (b)  
 (d) None of the above
- Q25.** How many true breeding pea plant varieties were selected by Mendel.  
 (a) 7 (b) 14  
 (c) 21 (d) 28
- Q26.** Experimental verification of the chromosomal theory of inheritance was done by.  
 (a) Morgan (b) Mendel  
 (c) Sutton (d) Boveri
- Q27.** Which of the following most appropriately describes haemophilia?  
 (a) Recessive gene disorder  
 (b) X - linked recessive gene disorder  
 (c) Chromosomal disorder  
 (d) Dominant gene disorder
- Q28.** A pleiotropic gene.  
 (a) controls multiple traits in an individual  
 (b) is expressed only in primitive plants  
 (c) controls a trait only in combination with another gene  
 (d) None of the above
- Q29.** Sickle cell anemia is.  
 (a) Characterized by elongated sickle like RBCs with a nucleus  
 (b) An autosomal linked dominant trait  
 (c) Caused by substitution of valine by glutamic acid in the beta globin chain of haemoglobin  
 (d) Caused by a change in a single base pair of DNA

- Q30.** Sex determination in humans takes place by.  
(a) sex chromosomes of father  
(b) measurement of sperm  
(c) measurement of ovum  
(d) sex chromosomes of mother

### ASSERTION AND REASONING

**Direction:** in the following questions, a statement of assertion (A) is followed by a statement of reason (R). Choose the correct option among a, b, c and d.

- Q1. Assertion (A):** Alfred Sturtevant, used the frequency of recombination, to measure the distance between genes.  
**Reason (R):** more frequency of recombination means, genes are located farther, low frequency of recombination means genes are located nearer.  
(a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
(b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
(c) Assertion (A) is true but reason(R) is false  
(d) Assertion (A) is false but reason(R) is true
- Q2. Assertion (A):** Experimental verification of the chromosomal theory of inheritance was done by Thomas Hunt Morgan.  
**Reason (R):** Morgan worked on butterflies.

- (a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
(b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
(c) Assertion (A) is true but reason(R) is false  
(d) Assertion (A) is false but reason (R) is true

- Q3. Assertion (A):** Characters are controlled by discrete units called factors.  
**Reason (R):** It is the statement of law of dominance.  
(a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
(b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
(c) Assertion (A) is true but reason(R) is false  
(d) Assertion (A) is false but reason (R) is true
- Q4. Assertion (A):** A single gene can exhibit multiple phenotypic expression. Such a gene is called a polygene.  
**Reason (R):** An example of polygene in humans is human skin colour.  
(a) Both assertion (A) and reason (R) are true and reason (R) is the correct explanation of assertion (A)  
(b) Both assertion (A) and reason (R) are true but reason (R) is not the correct explanation of assertion (A)  
(c) Assertion (A) is true but reason(R) is false  
(d) Assertion (A) is false but reason (R) is true

### SOLUTIONS MULTIPLE CHOICE

- S1. (b)** genetic is a broad field that deals with the inheritance of characteristics and the passing of characteristics from parents to offspring.
- S2. (c)** Mendel in the year 1856-1853 conducted experiments and proposed the laws of inheritance in living organisms.
- S3. (d)** Mendel selected 14 true-breeding pea plant varieties as pairs that were similar except for one character with contrasting traits for his experiment.
- S4. (c)** when Mendel self-pollinated the F<sub>1</sub> plants, he found that in F<sub>2</sub> generation some of the offerings were dwarf and some were tall, the ratio of tall to short was 3:1.
- S5. (b)** emasculation is the process of removal of anthers from a flower, the process is generally carried out during hybridization experiments.
- S6. (b)** the diagram is of a flower showing petal, stigma, anther, stamen and carpel.
- S7. (c)** in a cross between tall and a dwarf pea plant, the F<sub>1</sub> generations had tall and short pea plants in ratio of 1: 1 while in F<sub>2</sub> generation the ratio was 3:1.
- S8. (b)** in the F<sub>2</sub> generation there is one homozygous tall plant.
- S9. (b)** in the F<sub>2</sub> generation the genotypic ratio is 1:2:1.
- S10. (a)** the progenies of a test cross can be analysed easily to predict the genotype of the test organism.
- S11. (d)** the law of dominance is used to explain the expression of only one of the parental characteristics in a monohybrid cross in the F<sub>1</sub> and the expression of both in the F<sub>2</sub> generation.
- S12. (d)** Snapdragon shows incomplete dominance; it produces a flower that does not resembles any of the parent.
- S13. (b)** the genotypic ratio in incomplete dominance is exactly the same as in a Mendelian monohybrid cross.

- S14. (d)** Incomplete dominance results when a gene produces a less efficient enzyme or a non-functional enzyme or no enzyme at all.
- S15. (c)** The ABO blood group in humans is controlled by gene *I*. The plasma membrane of the RBC has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene.
- S16. (d)**  $I^A$  and  $I^B$  gene are completely dominant over *I*. If in a person genes  $I^A$  and *i* are present, only  $I^A$  expresses, as  $I^A$  shows complete dominance over *i*.
- S17. (c)** When Mendel crossed two plants differing in two pair of contrasting characters, the phenotypic ratio of the F2 generation was 9:3:3:1.
- S18. (c)** pleiotropy is a process in which a single gene can exhibit multiple phenotypic expressions.
- S19. (d)** Phenylketonuria is a disease that is caused by the mutation in the gene that codes for the enzyme phenyl alanine hydroxylase.
- S20. (b)** Henking could trace a specific nuclear structure all through spermatogenesis in a few insects and it was also observed by him that 50% of the sperm received the structure after spermatogenesis whereas the other 50% sperm did not receive it, he named this structure as X-body.
- S21. (b)** grasshopper is an example of XO type of sex determination in which the males have only one X-chromosome besides the autosomes whereas females have a pair of X-chromosomes.
- S22. (a)** female birds have two different sex chromosome, the female has one Z and one W chromosome while the male has a pair of Z chromosomes beside the autosome.
- S23. (a)** Humans have 23 pairs of chromosomes, 22 are exactly same in both males and females whereas the presence of X or Y is determinant of male or female.
- S24. (c)** 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).
- S25. (d)** In honey bee, development is different, the male develops without fertilisation via parthenogenesis into a male or a drone and the female via the fusion of male and female gametes.
- S26. (c)** In honeybee haploid-diploid sex determination system is present and has special characteristic features such as males produces sperms by mitosis.
- S27. (d)** Mutation is a change that occurs in a DNA sequence either due to mistakes or due to effect of smoking, sunlight or radiation.
- S28. (a)** mutations that occur in the somatic part of the body other than the germinal cells cannot be passed on to the descendants.
- S29. (d)** Mendelian disorders are transmitted to the offspring on the same lines as principles of inheritance and can be traced easily in a family by pedigree analysis.
- S30. (b)** the disease is caused due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour.
- S31. (b)** in pedigree analysis the inheritance of a particular trait is represented in the family tree over generation.
- S32. (c)** Mendelian disorders can either expresses themselves or can remain hidden, they can be either dominant or recessive.
- S33. (b)** colour blindness is a sex-linked recessive disorder and is caused due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour effect, it is due to mutation in certain genes present on X chromosome.
- S34. (a)** Males are more effected than the females because the genes that lead to red-green colour blindness are on the X-chromosome. Males have only one chromosome and females have two and the gene causing the disease is recessive.
- S35. (b)** The woman here is a carrier and carrier a recessive gene hence her son will have 50% chances of being colour-blind.
- S36. (c)** A female for haemophilia may transmit the disease to the sons, 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.
- S37. (c)** Sickle Cell anaemia is autosomal linked recessive trait that can be transmitted from parents to offspring when both the partners are carrier for the gene.
- S38. (c)** in Sickle Cell anaemia, heterozygous individuals are affected but they are carrier of the disease as there is 50% probability of transmission of the mutant gene to the progeny, only the homozygous individuals will show the diseased phenotype.
- S39. (b)** the substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.

- S40. (c)** Elongated sickle like Shaped RBC are unable to carry oxygen effectively.
- S41. (c)** in the disease, the affected individual lacks an enzyme that converts the amino acid phenylalanine to tyrosine.
- S42. (c)** in beta thalassaemia production of beta globin chain is affected, beta thalassaemia is controlled by a single gene HBB on chromosome 11 of each parent and occurs due to mutation of one or both the genes.
- S43. (b)** The disorder was first described by Langdon Down in the year 1866. It is trisomy in chromosome 21.
- S44. (c)** The individuals affected with this have 45 chromosomes, XO.
- S45. (b)** The visible characters are termed as phenotype.
- S46. (d)** Persons having Down's syndrome show trisomy of 21<sup>st</sup> chromosomes.
- S47. (c)** recombination is suppressed in the presence of linkage thus it promotes the appearance of more parental phenotypes.
- S48. (c)** Morgan gave the term 'linkage' while his experiments on *Drosophila*.
- S49. (b)** Crossing over is seen in prophase I during meiosis before the tetrads get aligned along the equator during metaphase I.
- S50. (d)** Mendel conducted hybridisation experiments on pea plant and took into account 7 pair of contrasting characters, crossed plants to obtain F<sub>1</sub>, F<sub>2</sub> and subsequent generations and thus proposed the above three laws.

### ASSERTION AND REASONING

- S1. (a)** Human skin colour is another classic example for this. In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive.
- S2. (c)** In human female, the sex chromosome is XX but the sex of the child is only determined by the male parent.
- S3. (d)** Down's Syndrome is a genetic disorder is caused due to the presence of an additional copy of the chromosome number 21 (trisomy of 21).
- S4. (a)** Thalassemia is 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 ( $\alpha$  and  $\beta$  chains) that make up haemoglobin.

### TRUE AND FALSE

- S1. (True)**
- S2. (False)** Human skin colour is classical example of polygenic inheritance.
- S3. (True)**
- S4. (False)** Phenylketonuria is an inborn error of metabolism.



## PRACTICE SOLUTIONS

- S1. (a)** Genetic variation is an important force in evolution because it allows natural selection to increase or decrease the frequency of alleles already in the population.
- S2. (b)** F1 always resembled to one of the parents and that was due to dominance.
- S3. (b)** The law of segregation states that two alleles for one gene do not blend with each other.
- S4. (a)** In Mendel's monohybrid cross the F<sub>2</sub> generations gives a ratio of 3:1
- S5. (d)** changed or modified allele can form three types of enzymes that is normal enzyme, non-functional enzyme or no enzyme at all.
- S6. (a)** modified allele is almost similar to normal allele and forms the same enzyme and produces the same phenotypic trait.
- S7. (c)** both can be possible, either there is no enzyme at all or the enzyme is non-functional is responsible for recessive trait.
- S8. (b)** The number of gametes produced will be  $= 2^n = 2^4 = 16$
- S9. (b)** the cross will be as follows:

	TY	Ty	tY	ty
TY	TTYT Tall, yellow	TTYy Tall, yellow	TtYY Tall, yellow	TtYy Tall, yellow
Ty	TTYy Tall, yellow	Ttyy Tall, green	TtYy Tall, yellow	Ttyy Tall, green
tY	TtYY Tall, yellow	TtYy Tall, yellow	ttYY Dwarf, yellow	ttYy Dwarf, yellow
ty	TtYy Tall, yellow	Ttyy Tall, green	ttYy Dwarf, yellow	ttty Dwarf, green

- S10. (c)** Both statistics and maths were first time applied in biology by Mendel during his studies.
- S11. (a)** in majority of insects the mechanism of sex determination is of XO type.
- S12. (d)** heterogamety means different sex gametes. Male heterogamety is found in males (XY) and some insects like grasshoppers and bugs (XO).
- S13. (c)** Sutton united the knowledge of chromosomal segregation with Mendel's principle and called it the chromosomal theory of inheritance.

- S14. (c)** Henking observed a nuclear structure in the sperm of a few insects that were present in 50% of sperm produced by spermatogenesis. The remaining 50% of sperm did not have it. He named it 'X body'.
- S15. (c)** in insects with XO type of sex determination, all sperms have X-chromosomes besides autosomes.
- S16. (c)** birds show female heterogamety.
- S17. (c)** Pedigree analysis is the study of family history about the inheritance of a particular trait in a several generations of a family. Pedigree analysis is done in the study of human genetics because control crosses cannot be possible in human being and age of human is more.
- S18. (b)** Mendelian disorders are mainly determined by the alteration or mutation in the single gene. These mutations are transmitted to the offspring.
- S19. (c)** Down's syndrome is a chromosomal disorder.
- S20. (d)** The chromosomal disorders on the other hand are caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- S21. (c)** Mendel's experiment had a large sampling size, he considered seven contrasting characters to pea plant to study because small sample sizes are prone to deviations caused by chance.
- S22. (c)**
- S23. (b)** The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.
- S24. (a)** Incomplete dominance is a form of intermediate inheritance, in which one allele for a specific trait is not completely expressed over its paired allele. This results in a third phenotype in which the expressed physical trait is a combination of the dominant and recessive phenotypes.
- S25. (b)** Mendel (father of genetics) selected 14 true-breeding pea plant varieties, as pairs, which were similar except for one character with contrasting traits.
- S26. (a)** Experimental Verification of the Chromosomal Theory of Inheritance was done by Thomas Hunt Morgan

- S27. (a)** Haemophilia is a sex linked recessive disease, in this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual a simple cut will result in non-stop bleeding.
- S28. (a)** Pleiotropism is the phenomenon of multiple effects of a gene in which a single pair of genes influences more than one character at the same time.
- S29. (c)** sickle cell anemia is caused by a change in a single base pair of DNA (substitution of glutamic acid by valine at the 6<sup>th</sup> position of the beta globin chain of the haemoglobin molecule).
- S30. (a)** sex determination in human is by the sex chromosomes of father.

#### ASSERTION AND REASONING

- S1. (a)** Alfred Sturtevant 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.
- S2. (c)** Morgan worked on Fruitflies.
- S3. (a)** Mendel's law of dominance states that characters are controlled by discrete units called factors that occur in pairs. In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).
- S4. (d)** A single gene can exhibit multiple phenotypic expression. Such a gene is called a pleiotropy.