27 Principles of Inheritance and Variation

Trend Analysis NEET

	NEET	Remarks
Number of Questions from 2023-16	33	3-4 Questions every year
Weightage	4.5%	

			NEET	
Year	Topic Name	Concept Used	No. of Questions	Difficulty Level
2023	Inheritance of two genes/ Pleiotropy, Genetic disorder	Recombination/ Pleiotropy/ Pedigree analysis/ Chromosomal disorders	5	Difficult/ Average
2022	Mendel's Law of inheritance/Linkage and Recombination/ Sex Determination/ Mutation/Genetic Disorder	Law of Inheritance/Linkage/Sex Determination/Mendalian Disorder	6	Difficult/ Average
2021	Genetic disorder/ Inheritance of one gene	Sickle cell Anaemia/ Punett Square	2	Easy
2020	Mendel's law of Inheritance, Inheritance of one gene & Inheritance of two genes/ Mutation & Genetic Disorders	Co-dominance/ Chromosomal theory of Inheritance/ Monohybrid Cross/ Mendelian disorder	3	Average/ Difficult
2019	Mendel's law of Inheritance, Inheritance of one gene & Inheritance of two genes/ Sex Determination/ Mutation & Genetic Disorders	Genetic Map/Linkage and Recombination/ Incomplete Dominance/Sex determination patterns/ Chromosomal disorder	4	Average/ Difficult
2018	Mendel's law of Inheritance, nheritance of one gene & Inheritance of two genes/ Sex Determination/ Mutation & Genetic Disorders/ Sex Determination/ Mutation & Genetic Disorders	Co-dominance/ Law of Inheritance/ Linkage and Recombination/ Chromosomal disorder	2	Average/ Difficult
2017	Mendel's law of Inheritance, inheritance of one gene & Inheritance of two genes /Mutation & Genetic Disorders	Co-dominance /Monohybrid Cross/ Mendelian disorder/ Chromosomal disorder	5	Average/ Difficult
2016	Mendel's law of Inheritance, Inheritance of one gene & Inheritance of two genes/ Mutation & Genetic Disorders	Test cross/ Law of Inheritance/ Monohybrid Cross/ Mendelian disorder/Chromosomal disorder	6	Average/ Difficult

Topic 27.1: Mendel's law of Inheritance

- Genetics is the branch of biology that deals with inheritance and variation of characters from parents to offspring.
- Inheritance is the process by characters are passed on from the parent to offspring while variation is the degree by which progeny differs from parents.

Mendel's Law of Inheritance

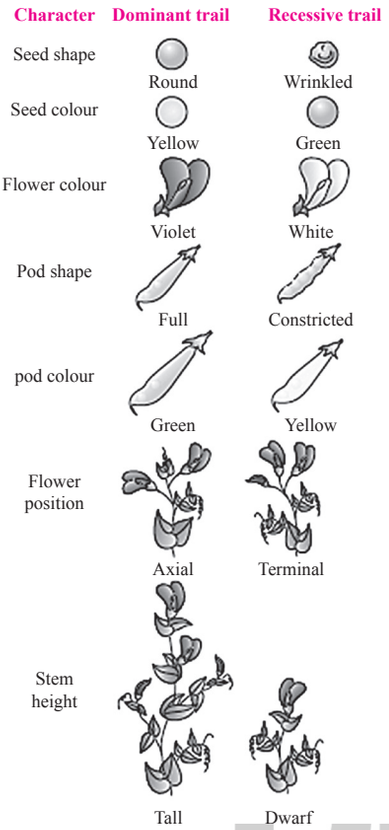


Fig. Seven pairs of contrasting traits in pea plant studied by Mendel

- Mendel was the first scientist to study the pattern of inheritance.
- He is known as the father of genetics and proposed the laws of inheritance.
- He selected pea plants (*Pisum sativum*) to carry out his study on the inheritance of characters from parents to offspring.

He selected pea plants because:

- Peas have many visible characters like tall/dwarf plants, round/wrinkled seeds, green/yellow pod etc.
- Peas have bisexual flowers and therefore they undergo natural self-pollination. Thus, pea plants produce offspring with same traits generation after generation.
- In pea plants, cross pollination can be easily achieved by emasculation in which the stamen of the flower is removed without affecting the pistil.
- Pea plants have short life span.
- They produce a large number of seed in one generation.

- *Mendel performed his experiments in three stages:*
  - Selection of pure or true breeding parents.
  - Hybridisation and obtaining first (F_1) generation.
  - Self-pollination of hybrid to get generations like F_2 and F_3 and so on.
- Mendel selected 7 pairs of true breeding pea varieties with easily distinguishable contrasting forms. The characters were:
- The true breeding variety produces offspring having similar characteristics. For example, white flower plant produced white flowered offspring etc.
- He obtained true breeding lines by continuous self-pollination for several generations.

Topic 27.2: Inheritance of One Gene (Monohybrid Cross)

Inheritance of One Gene

- Mendel selected true breeding tall (TT) and dwarf (tt) plants. Then he crossed these two plants. The seeds formed after fertilisation were grown and these plants that were formed represented the first filial or F_1 generation. All the F_1 plants obtained were tall.
- Then Mendel self-pollinated the F_1 plants and observed that all plants obtained in the F_2 generation were not tall. Instead one fourth of the F_2 plants were short.
- From this experiment Mendel concluded that the F_1 tall plants were not true breeding. They were carrying traits of both short height and tall height plants. They appeared tall only because the tall trait is dominant over the dwarf trait.

Mendel Observations

(i) The F_1 hybrid always showed one of the parental forms of the trait.

(ii) Both the parental forms of the trait (contrasting forms of the trait) appeared without any change in the F_2 generation.

(iii) The two contrasting forms in a trait did not show any blending either in the F_1 generation or in the F_2 generation.

(iv) The form of the trait that appeared in the F_2 hybrids is called dominant form and it appeared in the F_2 generation about three times in frequency as its alternate (recessive) form.

Mendel's proposition

- 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'.** Now a day we call them as **genes. Factors contain and carry hereditary information.**
- Gene is therefore are the **units of inheritance.**
- Genes which codes of a pair of contrasting traits are known as **alleles,** *i.e.* they are slightly different forms of the same gene.

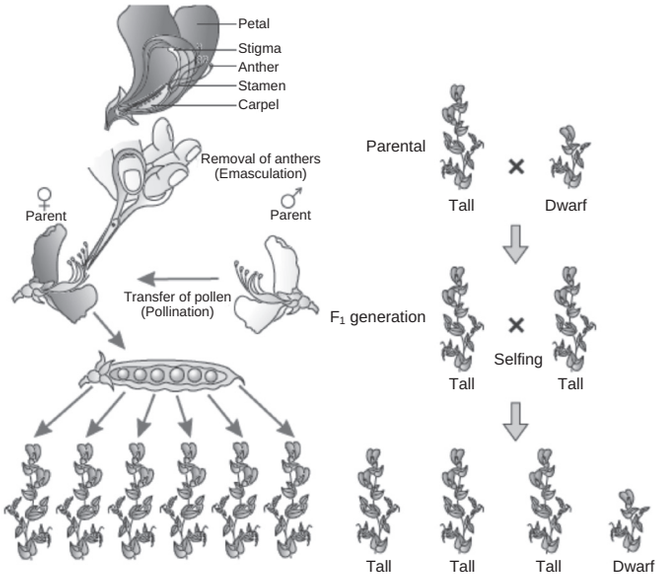


Fig.: Steps in making a cross in pea; Diagrammatic representation of monohybrid cross

Monohybrid Cross

- It is a cross between two plants, which differs in only one pair of contrasting characters. *e.g.* the cross between tall and dwarf pea plants
- *Steps involved in making a cross between tall and dwarf pea plants*
  - Selection of two pea plants with contrasting characters, say tall and dwarf pea plant.
  - Removal of anthers (emasculation) of one plant to avoid self-pollination. This is the female parent.
  - Collection of pollen grains from the other plant (male parent), which is then transferred to female parent for pollination.
  - Collection of seeds and production of offspring.

Parent Tall (Pure) × Dwarf (Pure)

↓

F_1-Generation All tall (Impure)

↓ Self pollination

Tall | Dwarf (Phenotypic ratio or basic ratio or Mendelian ratio)

F2-Generation 3 : 1

1 tall (pure) Sefling ↓ | 2 tall (impure) Sefling ↓ | 1 Dwarf (pure) Sefling ↓

F_3- Generation

All tall | 3 tall : 1 dwarf | All dwarf

Genotype ratio : Pure tall (1) : Impure tall (2) : Pure dwarf (1)

Mendelian ratio : Tall (3) : Dwarf (1)

Studying the cross

- TT, tt or Tt are genotypes while the traits, tall and dwarf are phenotypes.
- The capital letter T stands for tall trait while small letter t stands for dwarf trait.
- Even if a single "T"is present in the genotype, phenotype is tall. When "T" and "t"are present together, T dominates and suppresses the expression of "t". Therefore, T (for tallness) is dominant trait while t (for dwarfness) is recessive trait.
- TT and tt are homozygous while Tt is heterozygous.
- From the cross, it can be found that alleles of parental pair segregate from each other and only one allele is transmitted to the gamete.
- Gametes of TT will have only T alleles, gametes of tt will have only t alleles, but gametes of Tt will have both T and t alleles.

Punnett Square

- It is the graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross.
- Possible gametes are written on two sides, usually at top row and left columns, and combinations are represented in boxes.
- With the help of Punnett square, genotypic ratio in F_2 generation can be found.
- From the above punnet square, it is evident that genotypic ratio TT:Tt:tt is 1:2:1
- T ratio 1:2:1 or ¼ : ½ : ¼ of TT:Tt:tt can be derived from binomial expression $(ax + by)^2$
- Gamete bearing genes are in equal frequency of ½
- Hence, the expression can be written as

$(½\ T + ½\ t)\ 2 = (½\ T + ½\ t)\ (½\ T + ½\ t)$

$= ¼\ TT + ¼\ Tt + ¼\ Tt + ¼\ tt$

$= ¼\ TT + ½\ Tt + ¼\ tt$

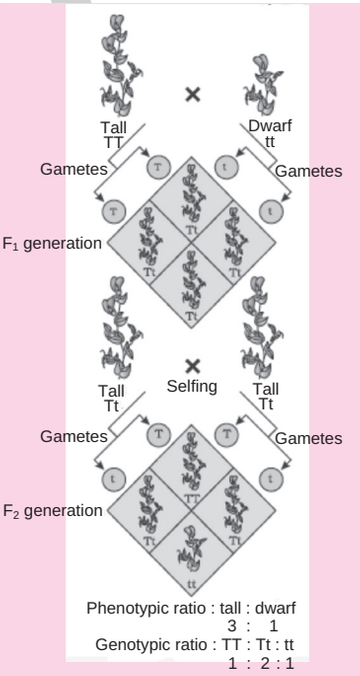


Fig.: A Punnett square used to understand a typical monohybrid cross conducted by Mendel between true-breeding tall plants and true-breeding dwarf plants **NEET 2016**

Back cross and Test cross

- **Back cross:** Back cross is the cross between an F_1 hybrid and any of its parent.

- **Test cross:** Test cross is a cross between an organism with unknown genotype and a recessive parent. It is used to determine whether the individual is homozygous or heterozygous for a trait.
- If the progenies produced by a test cross show 50% dominant trait and 50% recessive trait, then the unknown individual is heterozygous for the trait. On the other hand, if the progeny produced shows dominant trait, then the unknown individual is homozygous for a trait.
- The progeny of monohybrid test cross is 1:1 while the dihybrid tests cross is 1:1:1:1.

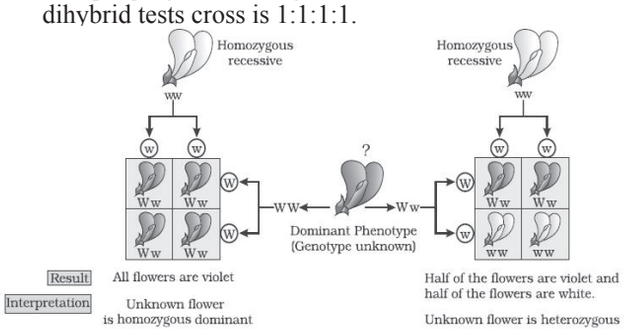


Fig.: Test Cross

- *Based on his experiments, Mendel proposed three laws of inheritance.*
  1. Law of dominance
  2. Law of segregation
  3. Law of independent assortment

1. First law - Law of dominance

- According to this law,
  - Characters are controlled by discrete units called factors, which occur in pairs.
  - It states that a dominant allele expresses itself in a monohybrid cross and suppresses the expression of recessive allele. However, this recessive allele for a character is not lost and remains hidden or masked in the progenies of F_1 generation and reappears in the next generation.
  - This law explains the expression of only one of the parental character in F_1 generation and expression of both in F_2 generation.
  - For *e.g.*, When pea plants with round seeds (RR) are crossed with plants with wrinkled seed (rr), all seeds in F_1 generation were found to be round (Rr). When these round seeds were self-fertilized, both the round and wrinkled seeds appeared in F_2 generation in 3:1 ratio. Hence, in F_1 generation the dominant characters (round seed) appeared and the recessive character (wrinkled seeds) got suppressed, which reappeared in F_2.

2. Second law- Law of segregation or Purity of gametes

- It states that "During gamete formation, the factors (alleles) of acharacter pair present in parents segregate from each othersuch that a gamete receives only one of the two factors".
- Homozygous parent produces all gametes that are similar i.e. they contain same type of allele while heterozygous parent produces two kinds of gametes (each having one allele) with equal proportion.

The concept of dominance

- In heterozygotes, there are dominant and recessive alleles.
- The normal (unmodified or functioning) allele of a gene produces a normal enzyme that is needed for the transformation of a substrate. The modified allele is responsible for production of
  1st case: The normal/less efficient enzyme or
  2nd case: A non-functional enzyme or
  3rd case: No enzyme at all
- **In the first case:** The modified allele is same as unmodified allele i.e. it produces the same phenotype/trait. The unmodified allele which represents the original phenotype is called the dominant allele.
- **In 2nd and 3rd cases:** The phenotype/trait is only dependent on the functioning of the unmodified allele. Here, the modified allele is generally termed as recessive allele.

Topic 27.3: Inheritance of Two Genes (Dihybrid cross)

Inheritance of Two Genes (Dihybrid Cross)

- **Dihybrid cross** is a cross between two parents, which differs in two pairs of contrasting characters.
- For *e.g.* cross between pea plants having round and yellow coloured seeds (RRYY) and wrinkled and green coloured seeds (rryy).
- In a dihybrid cross between two pea plants having round yellow and wrinkled green seeds, four types of gametes- RY, Ry, rY, ry, are produced.
- Each of these gamete segregate independent of each other, each having a frequency of 25% of the total gametes produced.
- On observing the F2, Mendel found that the yellow and green colour segregated in a 3:1 ratio. Round and wrinkled seed shape also segregated in a 3:1 ratio.

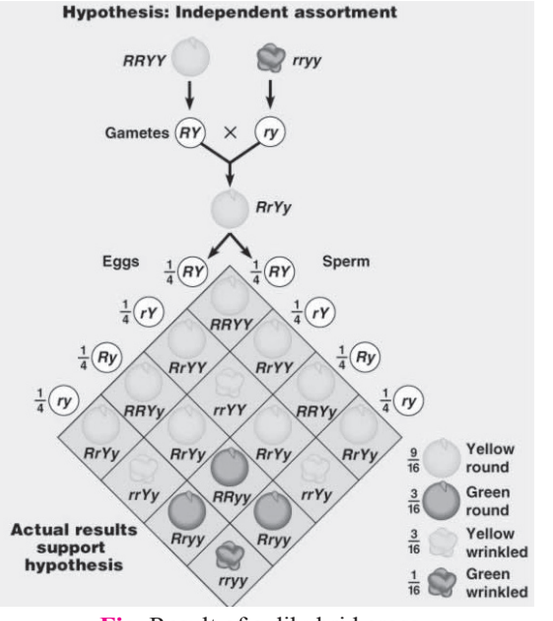


Fig. Result of a dihybrid cross.

- **Dihybrid phenotypic ratio**= 9:3:3:1
  Round yellow: Round green: Wrinkled yellow: Wrinkled green
  The ratio of 9:3:3:1 can be derived as a combination series of 3 yellow: 1 green, with 3 round: 1 wrinkled. i.e. **(3: 1) (3: 1) = 9: 3: 3: 1**
- Dihybrid genotypic ratio: 1:2:1:2:4:2:1:2:1
  RRYY =1 RRYy =2 RrYY =2
  RrYy =4 RRyy =1 Rryy =2
  rrYY =1 rrYy =2 rryy =1
  Based on the observations made on dihybrid cross, Mendel proposed a set of generalisation that we call Mendel's law of independent assortment.

Mendel's law of independent assortment:

- It states that 'when two pairs of traits are combined in a hybrid, segregation of one pair of characteristics is independent of the other pair of characters'.

Non-Mendelian Inheritance

(1) Incomplete Dominance

- When one of the two alleles of a gene is incompletely dominant over the other allele, it is termed as incomplete dominance.
- In this, neither of the alleles of a character is completely dominant over the other and the F_1 hybrid is intermediate between the two parents.
- For *e.g.* Flower colour in snapdrag on (dog flower or *Antirrhinum sp.*) and *Mirabilis jalapa* (4'O clock plant).
  - A monohybrid cross between the plants having red flowers and white flowers in *Antirrhinum* species will result in all pink colour flowers in the F_1 generation.
  - The progeny obtained in F_1 generation does not resemble either of the parents and exhibit intermediate characteristics. This is because, the dominant allele R, is partially dominant over the other allele, r.
  - Therefore, the recessive allele, r, also gets expressed in the F_1 generation resulting, in the production of intermediate pink flowering progenies with Rr genotypes.
  - Here, phenotypic and genotypic ratios are same.
    Phenotypic ratio= 1 Red: 2 Pink: 1 White
    Genotypic ratio= 1 (RR):2 (Rr):1(rr)
  - This means that **R** was not completely dominant over **r**.

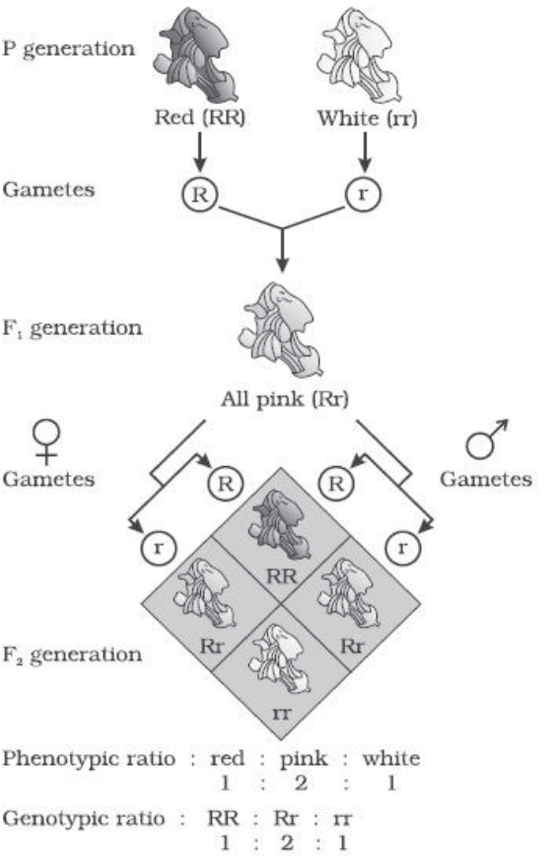


Fig.: Results of monohybrid cross in the plant Snapdragon, where one allele is incompletely dominant over the other allele.

2. Co-dominance

- When two alleles of a gene are equally dominant and express themselves even when they are together, are known as co-dominance.
- *E.g.* ABO blood grouping in human.
- ABO blood groups are controlled by the gene ***I***. Theplasma membrane of the RBC has sugar polymers thatprotrude from its surface and is controlled by the gene.
- The gene ***(I)*** has three alleles I_A, I_B and ***i***. The alleles I_A and I_B produce a slightly different form of the sugar whileallele ***i***doesn't produce any sugar.
- Each person possesses any two of the three I gene alleles.
- I_A and I_B are completely dominant over i, but when I^A and I^B are present together they both express their own types of sugars, i.e. they are co-dominant.
- Hence red blood cells have both A and B type sugars.

Alleles from parent I	Alleles from parent II	Genotype of offspring	Blood types (Phenotypes)
I^A	I^A	I^AI^A	A
I^A	I^B	$I^A I^B$	AB
I^A	i	I^Ai	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	I^BI^B	B
I^B	I	I^Bi	B
i	i	ii	O

Crosses of blood group (CO DOMINANCE)		
Blood group	**Possible genotype**	**Possible phenotype**
A X A	I^AI^A X I^AI^A	A
	I^AI^A X I^Ai	A
	I^Ai X I^Ai	A : O
B X B	I^BI^B X I^BI^B	B
	I^BI^B X I^Bi	B
	I^Bi X I^Bi	B; O
AB X AB	I^AI^B X I^AI^B	AB: A; B
O X O	ii X ii	O

3. Multiple allelism

- When a gene exists in more than two allelic forms, it shows the phenomenon of multiple allelism. *E.g.* ABO blood grouping. Gene for blood group exist in three allelic forms,I, I^B and i.

4. Pleiotropy

- It is the ability of a gene to have multiple phenotypic effects as it influences a number of characters simultaneously.
- The gene that has a multiple phenotypic effect because of its ability to control expression of a number of characters is called pleiotropic gene.
- For *e.g.* in Garden Pea, the gene which controls the flower colour also controls the colour of seed coat and presence of red spot in the leaf axil.

Chromosomal Theory of Inheritance

- *Mendel's work remained unrecognized for several years because of the following reasons.*
  - Lack of communication.
  - His mathematical approach to explain biological phenomenon was new and unacceptable to many of the biologists at that time.
  - His concept of **genes (factors)** as stable and discrete units was not accepted in the light of variations occurring continuously in nature, by his contemporaries.
  - Mendel could not provide any physical proof for the existence of factors.

Rediscovery of Mendel Results

- In 1900, three scientists namely, de Vries, Correns and von Tschermak independently rediscovered Mendel's results on the inheritance of character.
- **Chromosomal Theory of inheritance (1902):**
  - It was proposed by Walter Sutton and Theodore Boveriin 1902.
  - They discovered that behaviour of chromosomes was parallel to behaviour of genes.
  - Chromosome and genes both occur in pair. The two alleles of a gene pair are located on homologous sites of homologous chromosomes.
  - They further proposed that the pairing and separation of a pair of chromosomes lead to segregation of a pair of factors they carried.
  - Sutton united chromosomal segregation with Mendelian principles and proposed chromosomal theory of inheritance. It states that,
    - Chromosome and genes are present in pairs in diploid cells.
    - Homologous chromosomes separate during gamete formation (meiosis)
    - Fertilization restores the chromosome number to diploid condition.
    - The chromosomal theory of inheritance claims that, it is the chromosomes that segregate and assort independently.

Experimental verification of chromosomal theory

- **Thomas Hunt Morgan** formulated chromosomal theory of inheritance using fruit flies (***Drosophila melanogaster)***.
- *He chose Drosophila as his experimental model because,*
  - It breeds very quickly
  - They complete their life cycle in about two weeks. Therefore, breeding can be done throughout the year.
  - A single mating could produce a large number of progeny flies.
  - They can be grown on simple synthetic medium in the laboratory.
  - Easily distinguishable male and female
  - Hereditary variations that can be clearly seen with low power microscopes.

Morgan's experiment

- Morgan carried out several dihybrid crosses in ***Drosophila*** to study sex-linked genes. *E.g.*
  - **Cross 1:**
    Yellow-bodied, white-eyed females X Brown-bodied, red-eyed males (wild type)
  - **Cross 2:**
    White-eyed, miniature winged X Red eyed, large winged (wild type)
- Their F_1 progenies were obtained which were inter-crossed
- Then he obtained F_2 progeny and F_2 ratio was observed
- He found that
  - The two genes did not segregate independently of each other and the F_2 ratio deviated from the 9:3:3:1 ratio, (expected when the two genes are independent).

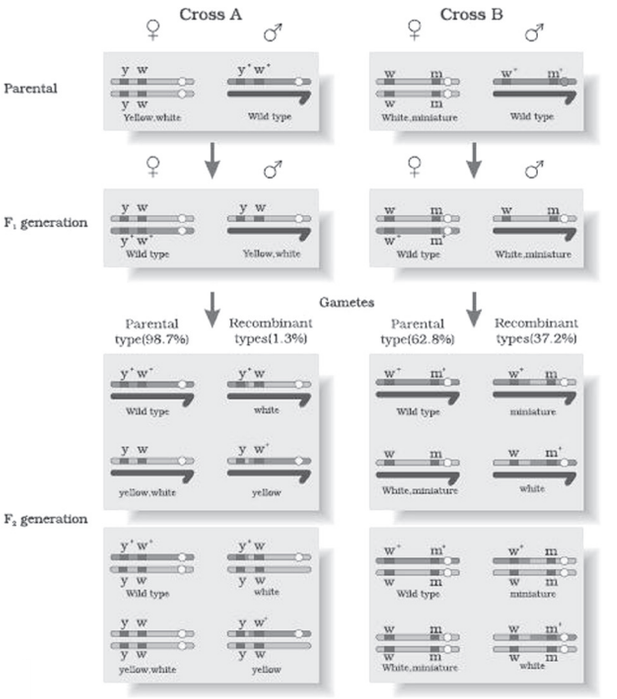


Fig.: Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript Note: The strength of linkage between y and w is higher than w and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

- ***Explanation of deviation from Mendelian ratio:***
  - Genes involved are located on the X chromosome.
  - When two genes are located on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type. Morgan attributed this due to the physical association or linkage of the two genes and coined the term **linkage.**
  - Morgan found that even when genes were grouped on the same chromosome, some genes were very **tightly linked** while others were **loosely linked.**
  - Tightly linked genes show low recombination. Looselylinked genes show high recombination.
  - Genes white and yellow were very tightly linked and showed only 1.3% recombination while white and miniature wing showed 37.2% recombination, hence loosely linked.
- Alfred Sturtevant used the recombination frequency between gene pairs as a measure of physical distance between genes and 'mapped' their position on the chromosome.
- In this way, genetic maps were prepared, which are used today for genome sequencing projects as in Human Genome Project.

Parallelism of behaviour between Chromosomes and Mendelian factors:

(i) Both the chromosomes and Mendelian factor are transmitted from one generation to another in an unaltered form.

(ii) A trait is represented by only one Mendelian factor inside a gamete. A gamete similarly contains a single chromosome out of a pair of homologous chromosomes due to meiosis that occurs before the formation of gametes.

(iii) An offspring contains two chromosomes of each types that are obtained from two parents through their gametes that are involved in fusion and formation of zygote. It also contains two Mendelian factor for each character. The factors come from two different parents through their gametes.

Linkage and Recombination

- **Recombination:** It is the generation of non-parental gene combinations.
- **Linkage:** linkage is defined as the co-existence of two or more genes in the same chromosome. If the genes are situated on the same chromosome and lie close to each other, then they are inherited together and are said to be linked genes.
  - For example, a cross between yellow bodied and white eyes and wild type parent in a Drosophila will produce wild type and yellow white progenies. It is because yellow bodied and white eyed genes are linked. Therefore, they are inherited together in progenies.

Topic 27.4: Polygenic Inheritance/ Pleiotropy

- Mendel studies describe that traits that have distinct alternate forms such as flower colour which are either purple or white.
- Such traits are generally controlled by three or more genes and are thus called as **polygenic traits**. Involvement of multiple genes polygenic inheritance also takes into account the influence of environment.
- Human skin colour is another classic example for this.
- In a polygenic trait the phenotype reflects the contribution of each allele, the effect of each allele is additive.
- The effect of a gene on a single phenotype or trait.
- There are however instances where a single gene can exhibit multiple phenotypic expression. Such a gene is called a **pleiotropic gene**. AIPMT 2015
- The 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 27.5: Sex Determination

Sex Determination

Autosomes and Sex chromosomes (allosomes)

- Those chromosomes (X and Y) which are involved in the determination of sex of an individual are called sex chromosomes while the other chromosomes are called autosomes.
- Number of autosomes is same in males and females.
- **Henking (1891)** studied spermatogenesis in some insects and observed that 50 % of sperm received a nuclear structure after spermatogenesis, whereas other 50 % sperm did not receive it. Henking called this structure as the **X body** (later it is called as **X-chromosome)**.

(i) Sex determination in grasshopper (XX-XO type)

- Sex-determination in grasshopper is **XX-XO type.**
- Here, female is homogametic, i.e. XX (all gametes are with X chromosomes) while male is heterogametic, i.e. XO (Gametes with X and gametes without X.
- Egg fertilized with sperm (with 'X' chromosome) became female (22+XX).
- Egg fertilized with sperm (without 'X' chromosome) became male (22 + Xo)

(ii) Sex determination in insects and mammals (XX-XY type):

- Both male and female has same number of chromosomes.
- Female have autosomes and a pair of X chromosomes. (AA+ XX).
- Male have autosomes and one **large 'X' chromosome** and one very small '**Y-chromosomes. (AA+XY)**
- This is called **male heterogamety** and **female homogamety**..

(iii) Sex determination in birds (ZZ-ZW type):

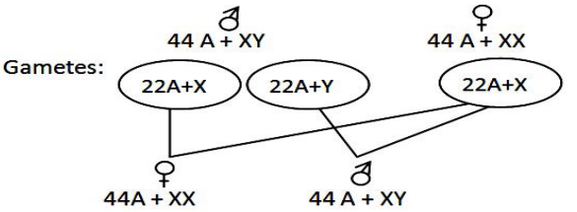
- Female birds have two different sex chromosomes designated as **Z** and **W.**
- Male birds have two similar sex chromosomes and called **ZZ.**
- Such type of sex determination is called **female heterogamety** and **male homogamety.**

(iv) Sex determination in honey bee:

- Sex determination in honey bee is based on the number of sets of chromosomes an individual receives.
- An offspring formed from the fertilization of a sperm and an egg developed into either queen (female) or worker (female).
- An unfertilized egg develops as a male (drone), by means of parthenogenesis.
- The male have half the number of chromosome than that of female.
- The female are diploid having 32 chromosomes and males are haploid i.e. having 16 numbers of chromosomes.
- This is called haplo-diploid sex determination system.
- Male produce sperms by mitosis.

(v) Sex Determination in humans (XX-XY type)

- Human has 23 pairs of chromosomes (22 pairs are autosomes and 1 pair is sex chromosomes).
- A pair of X-chromosomes (XX) is present in the female, whereas X and Y chromosomes are present in male.
- During spermatogenesis male produces two types of gametes. 50 % with X-chromosome and 50 % with Y-chromosome.
- Females produce only one ovum with an X-chromosome.
- There is an equal probability of fertilization of the ovum with the sperm carrying either X or Y chromosome.



- The sex of the baby is determined by the type of male gamete (X or Y) that fuses with the X chromosome of the female.

Topic 27.6: Mutation and Genetic disorder

Mutation

- It is a sudden heritable change in DNA sequence, which results in changes in the genotype and the phenotype of an organism.
- Mutation and Recombination are two phenomena, which leads to variation in DNA.
- **Frame-shift mutation:** Mutation which results due to loss (deletions) or gain (insertion/ duplication) of a DNA segment.
- **Point mutation**: Mutation that arise due to change in a single base pair of DNA, by substitution, deletion or insertion of a single nitrogenous base. *E.g.* sickle cell anaemia. It involves mutation in a single base pair in the beta globin chain of haemoglobin pigment in the blood. Glutamic acid in short arm of chromosome II gets replaced with valine at the sixth position.
- Mutation results in Chromosomal abnormalities (aberrations). Chromosomal aberrations are seen in **cancer cells.**

Mutagens

- Mutagens are agents that lead to mutations.
- It include,
  - **Physical mutagens:** UV radiation, α, β, γ rays, X-ray etc.
  - **Chemical mutagens:** Mustard gas, phenol, formalin, etc.

Pedigree Analysis

- Pedigree analysis is a record of occurrence of a trait in several generations of a family.
- It is based on the fact that certain characteristic features are heritable in a family, for e.g. eye colour, skin colour hair form and colour and other facial characteristics. Along with those features, there are other genetic disorders such as Mendelian disorders that are inherited in a family, generation after generation.
- Hence, by using pedigree analysis, for the study of specific traits or disorders, generation after generation, it is possible to trace the pattern of inheritance. In this, the inheritance of trait is represented as a tree, called family tree.
- The representation or chart showing family history is called **family tree (pedigree).**

Importance of pedigree analysis:

- It is useful for genetic counsellors to advice intending couples about the possibility of having children with genetic defects like haemophilia, thalassemia, etc.
- It is helpful to study certain genetic trait and find out the possibility of absence or presence of that trait in homozygous or heterozygous condition in a particular individual.
- In human genetics, pedigree study is utilized to trace the inheritance of a specific trait, abnormality or disease.

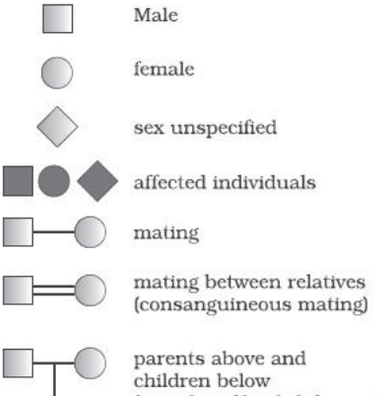


Fig.: Symbols used in the human pedigree analysis

Genetic Disorders

- A large number of diseases are known to be inherited from the parents to the offspring. Such diseases are known as genetic disorder.
- Most of these diseases are caused by expression of recessive allele.
- *Genetic disorder can be grouped into two categories: Mendelian disorders and Chromosomal disorders.*

1. Mendelian Disorders

- It is caused by alteration or mutation in the single gene.
- Their mode of inheritance follows the principles of Mendelian genetics.
- The pattern of inheritance of Mendelian disorders can be traced in a family by the pedigree analysis.
- *E.g.* Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalesemia, etc.
- ***Mendelian disorders can be:***
  - Autosomal dominant: *E.g.* Muscular dystrophy.
  - Autosomal recessive: *E.g.* Sickle cell anaemia, Albinism
  - Sex linked: *E.g.* Haemophilia.
- By pedigree analysis one can easily understand whether the trait is dominant or recessive.

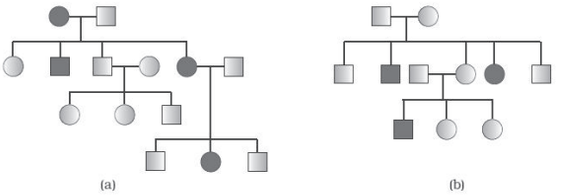


Fig.: Pedigree analysis of (a) Autosomal dominant trait (*E.g.* Myotonic dystrophy) (b) Autosomal recessive trait (*E.g.* Sickle-cell anaemia).

(i) Haemophilia (Royal disease):

- It is a sex linked recessive disease.
- It is transmitted from unaffected carrier female to their sons (male progeny).
- In this 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 no-stop bleeding.
- The heterozygous female (Carrier) for haemophilic may transmit the disease to sons.
- The possibility of a female becoming a haemophilic is very rare because mother of such female has to be at least carrier and father should be haemophilic.
- It is called royal disease because it shows a number of haemophilic descents as Queen Victoria was a carrier of the disease.

(ii) Sickle-cell anaemia:

- This is an autosome linked recessive disorder.
- It can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous).
- It is caused by point mutation in the beta-globin chain of haemoglobin pigment of the blood.
- The disease is controlled by a pair of allele, Hb^A and Hb^S.
  - Homozygous dominant (Hb^AHb^A): Normal
  - Heterozygous (Hb^AHb^S):Carrier of this disease.
  - Homozygous recessive (Hb^SHb^S): Affected (Diseased)
- The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the β- globin chain of the haemoglobin (Hb).
- This is due to the single base substitution at the sixth codon of the β-globin gene from GAG to GUG.
- The mutant Hb moleculeso formed undergoes polymerization under low oxygen tension causing the change in shape of the RBC from biconcave disc to elongated sickle like structure.

(iii) Phenylketonuria:

- It is an inborn error of metabolism.
- It is an autosomal recessive disease.
- The affected individual lacks an enzyme ***(phenylalanine hydroxylase)*** that converts the amino acid ***phenylalanine*** into ***tyrosine.*** As a result, phenylalanine accumulates andconverts into ***phenyl pyruvic acid*** and other derivatives.
- This acid accumulates in brain and leads to mental retardation.
- Phenyl pyruvic acid also gets excreted through urine since, kidneys poorly absorb it.

(iv) Thalassemia

- Thalassemia is an autosomal recessive disorder of the red blood cells.
- In Thalassemia one of the components of the haemoglobin molecule is inadequately produced or not produced at all.
  - If there is lack of α- chain production then the result is known as α-thalassemia.
  - If the component that is lacking is the β-chain, then the resulting condition is β- thalassemia.
- The reason for the inadequate or non- production of these components is a change in the genetic code (mutation), in that part of the DNA, which is the template for the production of the protein.
- A mutation may exist on one chromosome of a pair. The protein produced by the one, "healthy", and chromosome is enough to keep the individual well, even though his/her red cells are smaller than normal. Such an individual is known as a carrier (or heterozygote). A carrier may give his/her offspring either the healthy chromosome or the one bearing the mutation.
- Severe thalassaemia (Thalassemia Major) will result if a child inherits the abnormal (mutation bearing) chromosome from both parents. In other words both parents must be carriers if a major Thalassaemia disorder is present in the child. This situation is known as homozygous thalassemia.

2. Chromosomal disorders

- They are caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- It is of two types:

(a) **Aneuploidy**: The phenomenon ofgain or loss of one or more chromosomes due to failure of segregation of homologous pair of chromosomes during meiosis.

(b) **Polyploidy (Euploidy):** It is an increase in a **whole** set of chromosomes due to failure of cytokinesis after telophasestage of cell division. This is often seen in plants.

Examples for chromosomal disorders

(i) **Down's syndrome (Mongolism)**:
Cause: Presence of an additional copy of chromosome number 21 **(trisomy of 21).**

Genetic constitution: **45 A + XX or 45 A + XY** (*i.e.* **47** chromosomes).

Features of affected individual:

- They are short statured with small round head.
- Broad flat face.
- Furrowed big tongue and partially open mouth.
- Many "loops" on finger tips.
- Palm is broad with characteristic palm crease.
- Retarded physical, psychomotor and mental development.
- Congenital heart disease.

(ii) Klinefelter's syndrome:

Cause: Presence of an additional copy of X-chromosome in male.

Genetic constitution: **44 A + XXY** (*i.e.* **47** chromosomes).

Features of affected individual:

- Overall masculine development, however, the feminine development is also expressed. *E.g.*
  - Development of breast **(Gynaecomastia).**
  - Sterile.
  - Mentally retarded.

(iii) Turner's syndrome:

Cause: Absence of one ofthe X chromosomes in female.

Genetic constitution: **44 A + XO** (*i.e.* **45** chromosomes).

Features of affected female:

- Sterile as their ovaries are rudimentary.
- Lack of other secondary sexual characters.
- Dwarf.
- Mentally retarded.

Key Term Refresher – FIB

1. _____, conducted hybridisation experiments on garden peas for seven years (1856-1863) and proposed the laws of inheritance in living organisms.
2. Mendel selected _____ true-breeding pea plant varieties, as pairs which were similar except for one character with contrasting traits.
3. _____, are the units of inheritance.
4. TT and tt are called the _____ of the plant.
5. Hybrids contain alleles which express contrasting traits, the plants are _____.
6. Characters are controlled by discrete units called _____.
7. _____ can be found only when population studies are made.
8. _____ argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried.
9. Morgan worked with the tiny fruit flies, _____.
10. _____ is a phenomenon which results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism.
11. analysis of traits in a several of generations of a family is called the _____.
12. inborn error of metabolism is also inherited as the _____.
13. 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.
14. Failure of segregation of chromatids during cell division cycle results in the gain or loss of a chromosome(s), called _____.
15. The total number of chromosomes in a normal human cell is _____.

Concept Map

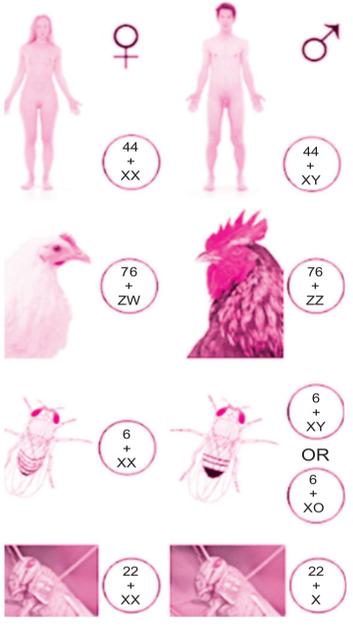
PRINCIPLES OF INHERITANCE AND VARIATIONS

- **Mendel's law of inheritance**
  - **Law of dominance**
    - According to this law, hybrid offspring will only inherit the dominant trait in phenotype
  - **Law of segregation**
    - Every individual possess 2 alleles and only 1 allele is passed on to the offspring
  - **Law of independent assortment**
    - Alleles of two different genes get sorted into gametes independently of one another.
- **Mutation**
  - Change in the DNA sequence of an organism
- **Pedigree analysis**

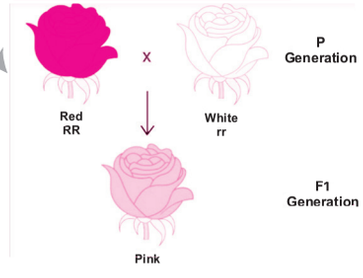
Normal female
Haemophilic female
Career female
Normal male
Haemophilic male

- **Sex-determination**

Different types of sex determination



- **Non-Mendalian Inheritance**
  - **Co-dominance**
  - **Incomplete dominance**



  - **Multiple alleles**

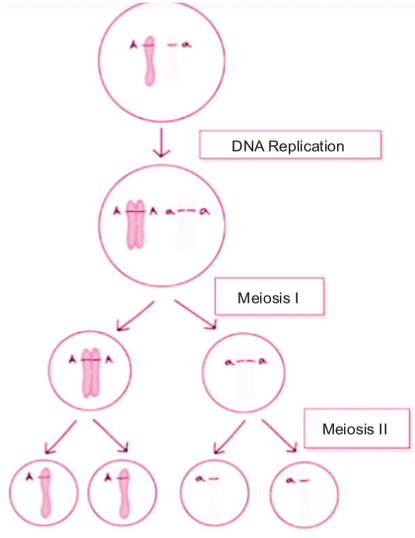
A	AB	A
AB	B	B
A	B	O

- **Linkage**
  - Tendency of genes closely positioned along a chromosome to segregate together at meiosis
- **Genetic disorders**

Disorders

. Down's syndrome (Trisomy of 21st chromosome)
. Turner's syndrome (XO)
. Klinerfelter's syndrome (XXY)
. Edward's syndrome (Trisomy of 18th chromosome)
. Sex-linked diseases

- **Chromosomal theory of inheritance**



Exercise 1 : NCERT Practice Problem Level-1

Mendel's law of Interitance

1. What contribute to the success of Mendel ?
   (a) Qualitative analysis of data
   (b) Observation of distinct inherited traits
   (c) His knowledge of biology
   (d) Consideration of one character at one time
2. The contrasting pairs of factors in Mendelian crosses are called
   (a) multiple alleles (b) allelomorphs
   (c) alloloci (d) paramorphs
3. Which one of the following traits of garden pea studied by Mendel was a recessive feature ?
   (a) Round seed shape (b) Axial flower position
   (c) Green seed colour (d) Green pod colour
4. The genes controlling the seven pea characters studied by Mendel are now known to be located on how many different chromosomes ?
   (a) Four (b) Seven (c) Six (d) Five
5. Suppose that in sheep, a dominant allele (B) produces black hair and a recessive allele (b) produces white hair. If you saw a black sheep, you would be able to identify
   (a) its phenotype for hair colour.
   (b) its genotype for hair colour.
   (c) the genotypes for only one of its parents.
   (d) the genotypes for both of its parents.
6. 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
7. Position of a gene on chromosome is called
   (a) locus (b) factor
   (c) cistron (d) nucleosome

Inheritance of One Gene

8. A gene is said to be dominant if
   (a) it expresses its effect only in homozygous state
   (b) it expresses its effect only in heterozygous condition
   (c) it expresses its effect both in homozygous and heterozygous condition
   (d) it never expresses its effect in any conditions
9. In hybridisation Tt × tt gives rise to the progeny of ratio 1999
   (a) 1 : 1 (b) 1 : 2 (c) 2 : 1 (d) 1 : 2 : 1
10. The genotype of a plant showing the dominant phenotype can be determined by :
    (a) test cross (b) dihybrid cross
    (c) pedigree analysis (d) back cross
11. A common test to find the genotype of a hybrid is by
    (a) crossing of one F_2 progeny with female parent
    (b) studying the sexual behaviour of F_1 progenies
    (c) crossing of one F_1 progeny with recessive parent
    (d) crossing of one F_2 progeny with male parent.
12. In order to find out the different types of gametes produced by a pea plant having the genotype AaBb, it should be crossed to a plant with the genotype:
    (a) AABB (b) AaBb (c) aabb (d) aaBB
13. Test cross in plants or in *Drosophila* involves crossing
    (a) between two genotypes with recessive trait
    (b) between two F_1 hybrids
    (c) the F_1 hybrid with a recessive genotype.
    (d) between two genotypes with dominant trait

Inheritance of Two Genes (Dihybrid cross)

14. Which Mendelian idea is depicted by a cross in which the F_1 generation resembles both the parents?
    (a) Law of dominance (b) Inheritance of one gene
    (c) Co-dominance (d) Incomplete dominance
15. Inheritances of skin colour in humans is an example of
    (a) point mutation (b) polygenic inheritance
    (c) codominance (d) chromosomal aberration
16. A pea plant with purple flowers is heterozygous for flower color. Its genotype is Pp. The P and p alleles in the pea plant's cells are located
    (a) next to each other on the same chromosome.
    (b) at the same location on homologous chromosomes.
    (c) on the X and Y chromosomes.
    (d) some distance apart on the same chromosome.
17. Lack of independent assortment of two genes A and B in fruit fly *Drosophila* is due to
    (a) repulsion (b) recombination
    (c) linkage (d) crossing over
18. Originally, genetic inheritance was thought to be a function of the blending of traits from the two parents. Which exception to Mendel's rules is an example of blending ?
    (a) Polygenic inheritance (b) Incomplete dominance
    (c) Codominance (d) Pleiotropism
19. A red-flowered plant crossed with a white-flowered plant of the same species, produced F_1 plants which all had pink flowers.
    Self-pollination of the F_1 plants produced and F_2 generation in which 39 plants had red flowers, 83 had pink flowers and 40 had white flowers. What does this experiment demonstrate ?
    (a) Codominance (b) Continuous variation
    (c) A dihybrid cross (d) Linkage
20. A girl has blood group A and her brother has blood group B. Which combination of genotypes cannot belong to their parents?

	Mother	Father
(a)	I^AI^A	I^BI^O
(b)	I^AI^B	I^AI^B
(c)	I^OI^O	I^AI^B
(d)	I^BI^O	I^AI^O

21. In Mendel's experiment 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 yellow cotyledons
(b) only wrinkled seeds with green cotyledons
(c) round seeds with yellow cotyledons, and wrinkled seeds with yellow cotyledons
(d) only round seeds with green cotyledons

22. A human male produces sperms with the genotypes AB, Ab, aB, and ab pertaining to two diallelic characters in equal proportions. What is the corresponding genotype of this person ?
(a) AaBB (b) AABb (c) AABB (d) AaBb

23. F_2 generation in a Mendelian cross showed that both genotypic and phenotypic ratios are same as 1 : 2 : 1. It represents a case of :
(a) Co-dominance
(b) Dihybrid cross
(c) Monohybrid cross with complete dominance
(d) Monohybrid cross with incomplete dominance

24. In blood group typing in human, if an allele contributed by one parent is I^A and an allele contributed by the other parent is i, the resulting blood group of the offspring will be
(a) A (b) B (c) AB (d) O

25. Across between parents with A and AB blood groups results in the offspring with which of the following blood groups?
(a) only A (b) only B
(c) A, B and AB (d) only O

Polygenic Inheritance/ Pleiotropy

26. Occasionally, a single gene may express more than one effect. The phenomenon is called
(a) multiple allelism (b) mosaicism
(c) pleiotropy (d) polygeny

Sex Determination

27. In human sex determination the key factor is
(a) Y-chromosome (b) Both (a) and (c)
(c) X-chromosome (d) None of these

28. Which one of the following conditions correctly describes the manner of determining the sex in the given example?
(a) Homozygous sex chromosomes (ZZ) determine female sex in birds.
(b) XO type of sex chromosomes determine male sex in grasshopper
(c) XO condition in human as found in Turner syndrome, determines female sex.
(d) Homozygous sex chromosomes (XX) produce male in *Drosophila.*

Mutation and Genetic disorder

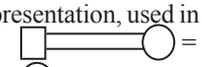
29. The exposure of X-rays enhances the frequency of
(a) linkage (b) crossing-over
(c) pairing of chromosomes (d) segregation

30. The gene of sickle cell anaemia is inherited by
(a) Blood cells (b) Bone cells
(c) Sex chromosomes (d) Autosomes

31. If a colour blind woman marries a normal visioned man, their sons will be
(a) one-half colour blind and one-half normal
(b) three-fourths colour blind and one-fourth normal
(c) all colour blind
(d) all normal visioned

32. The syndrome in which individual somatic cell contains three sex chromosomes XXY is called
(a) Klinefelter's syndrome (b) Turner syndrome
(c) Down's syndrome (d) Super female

33. 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) characterized by elongated sickle like RBCs with a nucleus
(d) an autosomal linked dominant trait

34. Which one of the following symbols and its representation, used in human pedigree analysis is correct?
(a)  = mating between relatives
(b)  = unaffected male
(c)  = unaffected female
(d)  = male affected

35. Mutations can be induced with :
(a) infra red radiations (b) I AA
(c) ethylene (d) gamma radiations

36. A person with unknown blood group under ABO system, has suffered much blood loss in an accident and needs immediate blood transfusion. His one friend who has a valid certificate of his own blood type offers blood donation without delay. What would have been the type of blood group of the donor friend?
(a) Type B (b) Type AB (c) Type O (d) Type A

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

38. Down's syndrome in humans is due to
(a) Two 'Y' chromosomes
(b) Three 'X' chromosomes
(c) Three copies of chromosome 21
(d) Monosomy

Exercise 2 : NCERT Practice Problem Level-2

Mendel's law of Interitance

1. It would have been very difficult for Mendel to draw conclusions about the patterns of inheritance if he had used cattle instead of peas. Why ?
(a) Cattle reproduce asexually.
(b) Cattle have small numbers of offspring.
(c) Cattle do not have observable phenotypes.
(d) Cattle do not have genotypes.

2. Mendel selected pea as material for his experiments because

(a) it is an annual plant with comparatively short life cycle.
(b) the flowers are self-pollinated.
(c) the number of seeds produced is quite large.
(d) all of the above.

Inheritance of One Gene

3. Why is the allele for wrinkled seed shape in garden peas considered recessive ?
(a) It "recedes" in the F_2 generation when homozygous parents are crossed.
(b) The trait associated with the allele is not expressed in heterozygotes.
(c) Individuals with the allele have lower fitness than that of individuals with the dominant allele.
(d) The allele is less common than the dominant allele.

4. The alleles found in haploid organisms cannot be dominant or recessive. Why ?
(a) Dominance and recessiveness describe interactions between two alleles of the same gene in the same individual.
(b) Because only one allele is present, alleles in haploid organisms are always dominant.
(c) Alleles in haploid individuals are transmitted like mitochondrial DNA or chloroplast DNA.
(d) Most haploid individuals are bacteria, and bacterial genetics is completely different from eukaryotic genetics.

5. Test cross involves
(a) crossing between two F_1 hybrids
(b) crossing the F_1 hybrid with a recessive genotype
(c) crossing between two genotypes with dominant trait
(d) crossing between two genotypes with recessive trait

6. The ratio of phenotypes in F_2 of a monohybrid cross is
(a) 3 : 1 (b) 1 : 2 : 1
(c) 9 : 3 : 3 : 1 (d) 2 : 1

Inheritance of Two Genes (Dihybrid cross)

7. 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 the above

8. In a particular plant, two genes control leaf shape and color. Round leaves (R) are dominant to jagged leaves (r). Yellow fruits (Y) are dominant to white fruits (y). A true-breeding round-leaved, yellow-fruited plant is mated with a jagged-leaved, white-fruited plant. What are the genotypes of the plants involved in this cross?
(a) RRYY × RRYY (b) RRYY × rryy
(c) RrYy × RrYy (d) RrYy × rryy

9. In the F_2 generation of a dihybrid cross between yellow, round seeded and green, wrinkled seeded pea plants, 17 out of 254 pea seeds were green and wrinkled. Other seeds were:
Yellow and round; green and round; yellow and wrinkled.
What do these results indicate ?
(a) Crossing over has occurred.
(b) Green and wrinkled are both recessive characters.
(c) The alleles for green and wrinkled are linked.
(d) The allele for green is recessive but not the allele for wrinkled.

10. Mendel's rules do not correctly predict patterns of inheritance for tightly linked genes or the inheritance of alleles that show incomplete dominance or epistasis. Does this mean that his hypothesis are incorrect ?
(a) Yes, because they are relevant to only a small number of organisms and traits.
(b) Yes, because not all data support his hypothesis.
(c) No, because he was not aware of meiosis or the chromosome theory of inheritance.
(d) No, it just means that his hypothesis are limited to certain conditions.

11. Which parental phenotypes would produce offspring with blood group phenotypes in the expected ratio of 1 type A : 1 type B ?

	Blood group of mother	*Blood group of father*
(a)	A	B
(b)	AB	AB
(c)	AB	B
(d)	AB	O

12. Mother and father of a person with '0' blood group have 'A' and 'B' blood group respectively. What would be the genotype of both mother and father.
(a) Mother is homozygous for 'A' blood group and father is heterozygous for 'B'
(b) Mother is heterozygous for 'A' blood group and father is heterozygous for 'B'
(c) Both mother and father are hetrozygous for 'A' and 'B' blood group respectively
(d) Both mother and father are homozygous for 'A' and 'B' blood group respectively.

13. Select the correct statement from the ones given below with respect to dihybrid cro**ss.**
(a) Tightly linked genes on the same chromosome show higher recombinations
(b) Genes far apart on the same chromosome show very few recombinations
(c) Genes loosely linked on the same chromosome show similar recombinations as the tightly linked ones
(d) Tightly linked genes on the same chromosome show very few recombinations

14. If two persons with 'AB' blood group marry and have sufficiently large number of children these children could be classified as 'A' blood group: 'AB' blood group: 'B' blood group in 1 : 2 : 1 ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of :
(a) incomplete dominance (b) Partial dominance
(c) Complete cominance (d) Codominance

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

Polygenic Inheritance/ Pleiotropy

16. Which one of the following is an example of polygenic inheritance ?

(a) Production of male honey bee
(b) Pod shape in garden pea
(c) Skin colour in humans
(d) Flower colour in *Mirabilis jalapa*

Sex Determination

17. *Drosophila* flies with XXY genotype are females but in case of humans, such individuals are abnormal males (Klinefelter's syndrome). This indicates that
(a) The Y chromosome has no role in sex determination
(b) In *Drosophila*, the Y chromosome is essential for sex determination
(c) The Y chromosome is male determining in humans
(d) The Y chromosome is female determining in *Drosophila*

Mutation and Genetic disorder

18. Which one of the followings is correctly matched with their chromosomal condition?
(a) Sickle cell anaemia – Heterozygous condition of Hbs gene
(b) Down's syndrome – Trisomy of chromosome 22
(c) Turner's syndrome – XO condition
(d) Klinefelter's syndrome – failure of cytokinesis after telophase

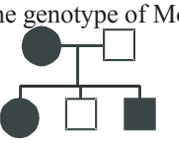
19. Which one of the following conditions in humans is correctly matched with its chromosomal abnormality/ linkage?
(a) Klinefelters syndrome – 44 autosomes + XXY
(b) Colour blindness –Y-linked
(c) Erythroblastosis foetalis - X-linked
(d) Downe syndrome – 44 autosomes + XO

20. 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) may be colour blind or may be of normal vision
(b) must be colour blind
(c) must have normal colour vision
(d) will be partially colour blind since he is heterozygous for the colour blind mutant allele

21. Phenylketonuria, Huntington's disease and sickle cell anaemia are caused respectively due to disorders associated with
(a) chromosome 7, chromosome 11 and chromosome 12
(b) chromosome 7, chromosome 12 and chromosome 11
(c) chromosome 4, chromosome 7 and chromosome 11
(d) chromosome 12, chromosome 4 and chromosome 11

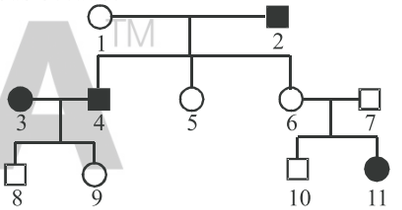
22. Which one is the incorrect statement with regards to the importance of pedigree analysis?
(a) It helps to trace the inheritance of a specific trait
(b) It confirms that DNA is the carrier of genetic information
(c) It helps to understand whether the trait in question is dominant or recessive
(d) It confirms that the trait is linked to one of the autosome

23. Given pedigree shows that the trait is inherited as autosomal dominant. Trace the genotype of Mother and Father



(a) Father AA, Mother aa
(b) Father AA, Mother Aa
(c) Father aa, Mother AA
(d) Father aa, Mother Aa

24. In Huntington's disease, the unaffected persons are homozygous for normal allele h. The following is erroneous because



(a) It shows both male and female affected by Huntingtons disease
(b) Either person 6 or 7 should have the disease, if individual 11 shows the disease.
(c) At least one of the 2 children (8, 9) should have the disease
(d) All of these

Exercise 3 : New Pattern Questions

Two Statement Type Questions

DIRECTION: *Read the statements carefully and answer the question on the basis of following options.*
(a) Both Statement I and Statement II are incorrect
(b) Statement I is correct but Statement II is incorrect
(c) Statement I is incorrect but Statement II is correct
(d) Both Statement I and Statement II are correct

1. **Statement I:** Frame-shift mutation arise due to change in single base pair of DNA.
Statement II: Point-mutation is due to insertion and deletion of base pair.

2. **Statement I:** Sutton and Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes.
Statement II: Sutton gave chromosomal theory of inheritance and he united the knowledge of chromosomal segregation with Mendalian principle of segregation.

3. **Statement I:** Sickle-cell anaemia is an autosome linked recessive trait that can be transmitted from parents.
Statement II: This disease is controlled by single pair of allele.

4. **Statement I:** Chromosomal aberrations are commonly observed in cancer cells.
Statement II: The alleration in chromosome results due to loss of a segment of DNA and gain of a segment of DNA.

5. **Statement I:** TT is the genotype of plant while tall is the phenotype.
Statement II: Hybrids contain alleles which express contrasting traits, the plant are heterozygous.

6. **Statement I:** Law of segregation is based on the fact that allele do not show any blading in F_1 generation.
Statement II: Snapdragon flower colar show co-dominance.

Assertion & Reason Questions

DIRECTION: *These questions consist of two statements, each printed as Assertion and Reason. While answering these questions, you are required to choose any one of the following four responses.*

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

7. **Assertion:** Human skin colour is controlled by 3 pairs of genes.
Reason: Skin colour in humans shows pleiotropic inheritance.

8. **Assertion:** In case of incomplete linkage, linked gene show new combination along with parental combination.
Reason: In case of incomplete linkage, linked genes are separated by crossing over.

9. **Assertion:** Aneuploidy may be of hypoploidy or hyperploidy (gain) type.
Reason: Monosomy lacks one pair of chromosomes.

10. **Assertion:** Cross of F_1 individual with recessive homozygous parent is a test cross.
Reason : No recessive individual is obtained in the monohybrid test cross.

11. **Assertion:** In *Antiorhinum*, selfing of F_1 pink-flowered plants produces same phenotypic and genotypic ratio.
Reason: Flower colour gene shows incomplete dominance.

12. **Assertion:** In honey bee drone have 16 chromosomes while queen has 32 numbers of chromosomes.
Reason: Male bees develop from unfertilised egg and female bees from fertilised eggs.

Four/Five Statement Type Questions

13. Which of the following statements are correct?
(i) Haemophilia is a sex-linked recessive disease.
(ii) Down's syndrome is due to aneuploidy.
(iii) Phenylketonuria is an autosomal dominant gene disorder.
(iv) Phenylketonuria is an autosomal recessive gene disorder.
(a) (i) and (iii) (b) (i), (ii) and (iii)
(c) (ii) and (iii) (d) (i), (ii) and (iv)

14. Which of the following statements are the correct?
(i) Failure of segregation of chromatids during cell division results in aneuploidy.
(ii) Chromosomal disorders are mainly determined by alteration or mutation in a single gene.
(iii) Thalasemia and cystic fibrosis are Mendelian disorders.
(iv) Sickle cell anemia is an X-linked trait.
(v) The thalassemia causes the formation of abnormal haemoglobin molecules resulting into anaemia.
(a) (i), (iii) and (v) (b) (i), (iii) and (iv)
(c) (iii), (iv) and (v) (d) (ii) and (iii)

15. Consider the following statement regarding linkage.
(i) The linked genes are located on the same chromosome.
(ii) Crossing over between linked gene is rare.
(iii) Linked gene are always inherited together.
(iv) Linked genes affect the percentage of homozygosity following hybridisation.
(v) Very tightly linked gene shows very low recombination.
Which of the following statement are correct?
(a) (i), (iii) and (v) (b) (i), (ii), (iii) and (v)
(c) (ii), (iii) and (iv) (d) (i), (iii) and (iv)

16. Which of the following statement is **not** correct for two genes that show 50% recombination frequency?
(i) The genes are tightly linked.
(ii) The genes show independent assortment.
(iii) If the genes are present on the same chromosome, they undergo more than one crossovers in every meiosis.
(iv) The genes may be on different chromosomes.
(a) Only (i) (b) Only (iii)
(c) (i) and (iv) (d) (i), (iii) and (iv)

Match the Following

17. Match column-I with column-II and select the correct answer using the codes given below.

Column-I		Column-II	
A.	ABO blood groups	I.	Dihybrid cross
B.	Law of segregation	II.	Monohybrid cross
C.	Law of Independent assortment	III.	Base pairs substitution
D.	Gene mutation	IV.	Multiple allelism

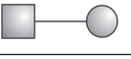
(a) A – II; B – I; C – IV; D – III
(b) A – IV; B – I; C – II; D – III
(c) A – IV; B – II; C – I; D – III
(d) A – II; B – III; C – IV; D – I

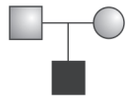
18. Match column-I with column-II and select the correct answer using the codes given below.

Column-I		Column-II	
A.	Turner syndrome	I.	Trisomy
B.	Linkage	II.	XX + XO
C.	Y-chromosome	III.	Morgan
D.	Down's syndrome	IV.	Testis determining factor

(a) A – II; B – I; C – IV; D – III
(b) A – II; B – I; C – IV; D – III
(c) A – IV; B – II; C – I; D – III
(d) A – II; B – III; C – IV; D – I

19. Match the symbols used in human pedigree analysis (given in column-I) with their name (given in column-II) and choose the correct option.

Column-I		Column-II	
A.		I.	Consanguineous mating
B.		II.	Normal female
C.		III.	Mating

D.		IV.	Parents with male child affected with disease
		V.	Sex unspecified

(a) A – III; B – I; C – II; D – IV
(b) A – II; B – I; C – IV; D – III
(c) A – III; B – V; C – I; D – IV
(d) A – III; B – I; C – V; D – IV

20. Match column-I with column-II and select the correct option from the codes given below.

Column-I		Column-II	
A.	Autosomal	I.	Down's syndrome recessive trait
B.	Sex-linked	II.	Phenylketonuria recessive trait
C.	Metabolic error	III.	Haemophilia linked to autosomal recessive
D.	Additional 21st	IV.	Sickle cell anaemia chromosome

(a) A – II; B – I; C – IV; D – III
(b) A – IV; B – I; C – II; D – III
(c) A – IV; B – III; C – II; D – I
(d) A – III; B – IV; C – I; D – II

21. Match the column I and II:

Column-I		Column-II	
A.	Pleiotropic gene	I.	Both allele express equally
B.	Co-dominance	II.	Change in nucleotide
C.	Epistasis	III.	One gene shows multiple phenotypic expression
D.	Mutation	IV.	Non - allelic gene inheritance

(a) A – I; B – II;, C – III; D – IV
(b) A – II; B – III; C – IV; D – I
(c) A – III; B – I; C – IV; D – II
(d) A – I; B – III; C – IV; D – II

Exercise 4 : NCERT Exemplar

1. All genes located on the same chromosome
(a) Form different groups depending upon their relative distance
(b) Form one linkage group
(c) Will not from any linkage group
(d) Form interactive group that affect the phenotype

2. Conditions of a karyotype 2n ±1 and 2n ± 2 are called
(a) aneuploidy (b) polyploidy
(c) Allopolyploidy (d) monosomy

3. Distance between the genes and percentage of recombination shows
(a) a direct relationship (b) an inverse relationship
(c) a parallel relationship (d) no relationship

4. If a genetic disease is transferred from a phenotypically normal but carrier female to only same of the male progeny, the disease is
(a) Autosomal dominant (b) autosomal recessive
(c) sex-linked dominant (d) sex-linked recessive

5. In sickle-cell anaemia glutamic acid is replaced by valine. Which one of the following triplet codes for valine ?
(a) G G G (b) AAG (c) GAA (d) GUG

6. Person having genotype $I^a I^b$ would show the blood group as AB. This is because of
(a) Pleiotropy (b) Codominance
(c) segregation (d) incomplete dominance

7. ZZ/ZW type of sex determination is seen in
(a) Platypus (b) Snails
(c) Cockroach (d) Peacock

8. Across between two tall plants resulted in offspring having few dwarf plants. What would be the genotypes of both the parents ?
(a) TT and Tt (b) Tt and Tt
(c) TT and TT (d) Tt and tt

9. In a dihybrid cross, If you get 9 : 3: 3: 1 ratio it denotes that
(a) the alleles of two genes are interacting with each other
(b) it is a multigenic inheritance
(c) It is a case of multiple allelism
(d) The alleles of two genes are segregating independently

10. Which of the following will not result in variations among siblings ?
(a) Independent assortment of genes
(b) Crossing over
(c) Linkage
(d) Mutation

11. Mendel's law of independent assortment holds good for genes situated on the
(a) non-homologous chromosomes
(b) homologous chromosomes
(c) extra nuclear genetic element
(d) Same chromosome

12. Occasionally, a single gene may express more than one effect. The phenomenon is called.
(a) multiple allelism (b) mosaicism
(c) pleiotropy (d) polygeny

13. In a certain taxon of insects some have 17 chromosomes and the others have 18 chromosomes. The 17 and 18 chromosome-bearing organisms are
(a) males and females respectively
(b) females and males, respectively
(c) all males
(d) all females

14. The inheritance pattern of a gene over generations among humans is studied by the pedigree analysis. Character studied in the pedigree analysis is equivalent to
(a) quantitative trait (b) Mendelian trait
(c) Polygenic trait (d) maternal trait

15. It is said that Mendel proposed that the factor controlling any character is discrete and independent. This proposition was based on the

(a) results of F_3-generation of a cross
(b) observations that the offspring of a cross made between the plants having two contrasting characters shown only one character without any blending
(c) Self-pollination of F_1 offsprings
(d) cross-pollination of F_1- generation with recessive parent

16. Two genes 'A' and 'B' are linked. In a dihybrid cross involving these two genes, the F_1- heterozygote is crossed with homozygous recessive parental type (aa bb). what would be the ratio of offspring in the next generation ?
(a) 1: 1 : 1: 1 (b) 9: 3 : 3: 1
(c) 3: 1 (d) 1: 1

17. In the F_2-generation of a Mendelian dihybrid cross the number of phenotypes and genotypes are
(a) phenotypes -4, genotypes-16
(b) phenotypes -9, genotypes -4
(c) Phenotypes-4, genotypes-8
(d) Phenotypes-4, genotypes-9

18. Mother and father of a person with '0' blood group have 'A' and 'B' blood group respectively. What would be the genotype of both mother and father.
(a) Mother is homozygous for 'A' blood group and father is heterozygous for 'B'
(b) Mother is heterozygous for 'A' blood group and father is heterozygous for 'B'
(c) Both mother and father are hetrozygous for 'A' and 'B' blood group respectively
(d) Both mother and father are homozygous for 'A' and 'B' blood group respectively.

Exercise 5 : Past Year Topic-Wise Questions

Mendel's law of Inheritance, Inheritance of one gene, Inheritance of two genes, Linkage and Recombination, Polygenic Inheritance, Pleiotropy

1. Frequency of recombination between gene pairs on same chromosome as a measure of the distance between genes to map their position on chromosome, was used for the first time by **2023**
(a) Thomas Hunt Morgan (b) Sutton and Boveri
(c) Alfred Sturtevant (d) Henking

2. The phenomenon of pleiotropism refers to **2023**
(a) Presence of several alleles of a single gene controlling a single crossover
(b) Presence of two alleles, each of the two genes controlling a single trait
(c) A single gene affecting multiple phenotypic expression
(d) More than two genes affecting a single character

3. Match List-I with List-II. **Ph-II 2023**

	List-I (Type of cross)		List-II (Phenotypic ratio)
(A)	Monohybrid Cross	(I)	1 : 1
(B)	Dihybrid Cross	(II)	1 : 2 : 1
(C)	Incomplete dominance	(III)	3 : 1
(D)	Test Cross	(IV)	9 : 3 : 3 : 1

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

4. A heterozygous pea plant with violet flowers was crossed with a homozygous pea plant with white flowers.
Violet is dominant over white. Which one of the following represents the expected combinations among 40 progenies formed? **Ph-II 2023**
(a) All 40 produced violet flowers
(b) All 40 produced white flowers
(c) 30 produced violet and 10 produced white flowers
(d) 20 produced violet and 20 produced white flowers

5. A certain plant homozygous for yellow seeds and red flowers was crossed with a plant homozygous for green seeds and white flowers. The F_1 plants had yellow seeds and pink flowers. The F_1 plants were selfed to get F_2 progeny. Assuming independent assortment of the two characters, how many phenotypic categories are expected for these characters in the F_2 generation? **Ph-II 2023**
(a) 4 (b) 6
(c) 9 (d) 16

6. Given below are two statements: **2022**
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:
(a) Both Statement I and Statement II are incorrect
(b) Statement I is correct but Statement II is incorrect
(c) Statement I is incorrect but Statement II is correct
(d) Both Statement I and Statement II are correct

7. Given below are two statements: one is labelled as **Assertion (A)** and the other is labelled as **Reason (R)**. **2022**
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 **most appropriate** answer from the options given below:
(a) Both (A) and (R) are correct but (R) is not the correct explanation of (A)
(b) (A) is correct but (R) is not correct
(c) (A) is not correct but (R) is correct
(d) Both (A) and (R) are correct and (R) is the correct explanation of (A)

8. The recombination frequency between the genes 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

9. The chromosomal theory of inheritance was proposed by
(a) Robert Brown (b) Thomas Morgan **Ph-II 2022**
(c) Sutton and Boveri (d) Gregor Mendel

10. What is the expected percentage of F_2 progeny with yellow and inflated pod in dihybrid cross experiment involving pea plants with green coloured, inflated pod and yellow coloured constricted pod? **Ph-II 2022**
(a) 9% (b) 100% (c) 56.25% (d) 18.75%

11. The production of gametes by the parents, formation of zygotes, the F_1 and F_2 plants, can be understood from a diagram called- **2021**
(a) Net square (b) Bullet square
(c) Punch square (d) Punnett square

12. Chromosomal theory of inheritance was proposed by
(a) Watson and Crick (b) Sutton and Boveri
(c) Bateson and Punnet (d) T.H. Morgan **Ph-II 2020**

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

14. The production of gametes by the parents, the formation of zygotes, the F_1 and F_2 plants, can be understood using
(a) Wenn diagram (b) Pie diagram **2019, Odisha**
(c) A pyramid diagram (d) Punnet square

15. In a marriage between male with blood group A and female with blood group B, the progeny had either blood group AB or B. What could be the possible genotype of parents?
(a) I^A i (Male) ; I^B i (Female) **2019, Odisha**
(b) I^A i (Male) ; I^B I^B (Female)
(c) I^A I^A (Male) ; I^B I^B (Female)
(d) I^A I^A (Male) ; I^B i (Female)

16. In *Antirrhinum* (Snapdragon), a red flower was crossed with a white flower and in F_1 generation, pink flowers were obtained. When pink flowers were selfed, the F_2 generation showed white, red and pink flowers. Choose the incorrect statement from the following. **2019**
(a) This experiment does not follow the principle of dominance.
(b) Pink colour in F_1 is due to incomplete dominance.
(c) Ratio of F_2 is $\frac{1}{4}$ (Red): $\frac{1}{2}$ (Pink): $\frac{1}{4}$ (White)
(d) Law of Segregation does not apply in this experiment.

17. What map unit (Centimorgan) is adopted in the construction of genetic maps? **2019**
(a) A unit of distance between two expressed genes, representing 10% cross over.
(b) A unit of distance between two expressed genes, representing 100% cross over.
(c) A unit of distance between genes on chromosomes, representing 1% cross over.
(d) A unit of distance between genes on chromosomes, representing 50% cross over

18. Which of the following characteristics represent 'Inheritance of blood groups' in humans? **2018**
(i) Dominance (ii) Co-dominance
(iii) Multiple allele (iv) Incomplete dominance
(v) Polygenic inheritance
(a) (ii), (iii) and (v) (b) (i), (ii) and (iii)
(c) (i), (iii) and (v) (d) (ii), (iv) and (v)

19. The genotypes of a husband and wife are I^AI^B and I^Ai. Among the blood types of their children, how many different genotypes and phenotypes are possible? **2017**
(a) 3 genotypes ; 4 phenotypes
(b) 4 genotypes ; 3 phenotypes
(c) 4 genotypes ; 4 phenotypes
(d) 3 genotypes ; 3 phenotypes

20. Among the following characters, which one was not considered by Mendel in his experiments on pea? **2017**
(a) Trichomes – Glandular or non-glandular
(b) Seed – Green or Yellow
(c) Pod – Inflated or Constricted
(d) Stem – Tall or Dwarf

21. Which one from those given below is the period for Mendel's hybridisation experiments? **2017**
(a) 1840 - 1850 (b) 1857 - 1869
(c) 1870 - 1877 (d) 1856 - 1863

22. A tall true breeding garden pea plant is crossed with a dwarf true breeding garden pea plant. When the F_1 plants were selfed the resulting genotypes were in the ratio of **2016**
(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

23. Match the terms in Column-I with their description in Column-II and choose the correct option. **2016**

Column-I	Column-II
(A) Dominance	(i) Many genes govern a single character
(B) Codominance	(ii) In a heterozygous organism, only one allele expresses itself
(C) Pleiotropy	(iii) In a heterozygous organism, both alleles express themselves fully
(D) Polygenic inheritance	(iv) A single gene influences many characters

	(A)	(B)	(C)	(D)
(a)	(ii)	(i)	(iv)	(iii)
(b)	(ii)	(iii)	(iv)	(i)
(c)	(iv)	(i)	(ii)	(iii)
(d)	(iv)	(iii)	(i)	(ii)

24. In a test cross involving F_1 dihybrid flies, more parental type offspring were produced than the recombinant type offspring. This indicates **2016**
(a) the two genes are located on two different chromosomes.
(b) chromosomes failed to separate during meiosis.
(c) the two genes are linked and present on the same chromosome.
(d) both of the characters are controlled by more than one gene.

25. The mechanism that causes a gene to move from one linkage group to another is called **Ph-II 2016**
(a) Translocation (b) Crossing-over
(c) Inversion (d) Duplication

26. How many pairs of contrasting characters in pea plants were studied by Mendel in his experiments? **2015 RS**
(a) Six (b) Eight (c) Seven (d) Five

27. Which is the most common mechanism of genetic variation in the population of sexually reproducing organism?
(a) Chromosomal aberrations **2015 RS**
(b) Genetic drift
(c) Recombination
(d) Transduction

28. Alleles are **2015 RS**
(a) true breeding homozygotes
(b) different molecular forms of a gene
(c) heterozygotes
(d) different phenotype

29. In his classic experiments on Pea plants, Mendel did not use **2015 RS**
(a) Pod length (b) Seed shape
(c) Flower position (d) Seed colour

30. A man with blood group 'A' marries a woman with blood group 'B'. What are all the possible blood groups of their offsprings? **2015 RS**
(a) A,B and AB only (b) A,B,AB and O
(c) O only (d) A and B only

31. Multiple alleles are present: **2015 RS**
(a) At different loci on the same chromosome
(b) At the same locus of the chromosome
(c) On non-sister chromatids
(d) On different chromosomes

32. A gene showing codominance has: **2015 RS**
(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

33. The term 'linkage' was coined by: **2015 RS**
(a) T. Boveri (b) G. Mendel
(c) W. Sutton (d) T.H. Morgan

34. A pleiotropic gene: **2015 RS**
(a) is a gene evolved during Pliocene.
(b) controls a trait only in combination with another gene
(c) controls multiple traits in an individual.
(d) is expressed only in primitive plants

35. Fruit colour in squash in an example of: **2014**
(a) Recessive epistasis (b) Dominant epistasis
(c) Complementary genes (d) Inhibitory genes

36. If two persons with 'AB' blood group marry and have sufficiently large number of children these children could be classified as 'A' blood group: 'AB' blood group: 'B' blood group in 1 : 2 : 1 ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of : **2013**
(a) incomplete dominance (b) Partial dominance
(c) Complete cominance (d) Codominance

37. Which Mendelian idea is depicted by a cross in which the F_1 generation resembles both the parents? **2013**
(a) Law of dominance (b) Inheritance of one gene
(c) Co-dominance (d) Incomplete dominance

38. Which of the following statements is not true of two genes that show 50% recombination frequency? **2013**
(a) The genes are tightly linked
(b) The genes show independent assortment
(c) If the genes are present on the same chromosome, they undergo more than one crossovers in every meiosis
(d) The genes may be on different chromosomes

39. Genetic variation in a population arises due to **Kar.2013**
(a) Mutations only
(b) Recombination only
(c) Mutations as well as recombination
(d) Reproductive isolation and selection

Sex Determination

40. Select the correct statement/s with respect to mechanism of sex determination in Grasshopper. **Ph-II 2023**
(A) It is an example of female heterogamety.
(B) Male produces two different types of gametes either with or without X chromosome.
(C) Total number of chromosomes (autosomes and sex chromosomes) is same in both males and females.
(D) All eggs bear an additional X chromosome besides the autosomes.
Choose the correct answer from the options given below:
(a) (A) only (b) (A) and (C) only
(c) (B) and (D) only (d) (A), (C) and (D) only

41. XO type of sex determination can be found in: **2022**
(a) Birds (b) Grasshoppers
(c) Monkeys (d) *Drosophila*

42. Match the items of Column-I with Column-II: **2020**

Column-I	Column-II
(A) XX-XO method	(i) Turner's syndrome of sex determination
(B) XX-XY method	(ii) Female heterogametic of sex determination
(C) Karyotype-45	(iii) Grasshopper
(D) ZW-ZZ method	(iv) Female of sex homogametic determination

Select the correct option from the following :
(a) (A)-(iv), (B)-(ii), (C)-(i), (D)-(iii)
(b) (A)-(ii), (B)-(iv), (C)-(i), (D)-(iii)
(c) (A)-(i), (B)-(iv), (C)-(ii), (D)-(iii)
(d) (A)-(iii), (B)-(iv), (C)-(i), (D)-(ii)

43. Select the incorrect statement. **2019**
(a) Male fruit fly is heterogametic.
(b) In male grasshoppers, 50% of sperms have no sex-chromosome.
(c) In domesticated fowls, sex of progeny depends on the type of sperm rather than egg.
(d) Human males have one, of their sex-chromosome much shorter than the other.

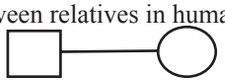
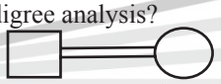
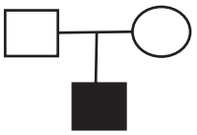
44. A woman has an X-linked condition on one of her X chromosomes. This chromosome can be inherited by **2018**
(a) Only daughters (b) Only sons
(c) Both sons and daughters (d) Only grandchildren

45. In our society women are blamed for producing female children. Choose the correct answer for the sex-determination in humans **2013**
(a) Due to some defect in the women
(b) Due to some defect like aspermia in man
(c) Due to the genetic make up of the particular sperm which fertilizes the egg
(d) Due to the genetic make up of the egg

Mutation, Genetic Disorders

46. Which of the following statements are correct about Klinefelter's Syndrome? **2023**
(i) This disorder was first described by Langdon Down (1866).
(ii) Such an individual has overall masculine development. However, the feminine developement is also expressed.
(iii) The affected individual is short statured.
(iv) Physical, psychomotor and mental development is retarded.
(v) Such individuals are sterile.
Choose the correct answer from the options given below:
(a) (i) and (ii) only (b) (iii) and (iv) only
(c) (ii) and (v) only (d) (i) and (v) only

47. Broad palm with single palm crease is visible in a person suffering from– **2023**
(a) Down's syndrome (b) Turner's syndrome
(c) Klinefelter's syndrome (d) Thalassemia

48. Which one of the following symbols represents mating between relatives in human pedigree analysis? **2023**
(a) 
(b) 
(c) 
(d) 

49. In which disorder change of single base pair in the gene for beta globin chain results in change of glutamic acid to valine? **PH-II 2023**
(a) Haemophilia (b) Phenylketonuria
(c) Thalassemia (d) Sickle cell anaemia

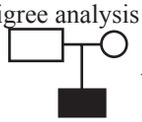
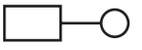
50. Select the correct statements about sickle cell anaemia. **PH-II 2023**
(A) There is a change in gene for beta globin.
(B) In the beta globin, there is valine in the place of Lysine.
(C) It is an example of point mutation.
(D) In the normal gene U is replaced by A.
Choose the correct answer from the options given below:
(a) (A), (B) and (D) only (b) (A) and (C) only
(c) (B), (C) and (D) only (d) (B) and (D) only

51. Which of the following occurs due to the presence of autosome linked dominant trait? **2022**
(a) Myotonic dystrophy (b) Haemophilia
(c) Thalessemia (d) Sickle cell anaemia

52. If a colour blind female marries a man whose mother was also colour blind, what are the chances of her progeny having colour blindness?
(a) 50% (b) 75% (c) 100% (d) 25% **2022**

53. Given below are two statements :
Statement I: Sickle cell anaemia and Haemophilia are autosomal dominant traits.
Statement II: Sickle cell anaemia and Haemophilia are disorders of the blood.
In the light of the above statements, choose the correct answer from the options given below: **PH-II 2022**
(a) Statement is incorrect but Statement II is correct
(b) Both Statement I and Statement II are correct
(c) Both Statement I and Statement II are incorrect
(d) Statement I is correct but Statement II is incorrect

54. If a female individual is with small round head, furrowed tongue, partially open mouth and broad palm with characteristic palm crease. Also the physical, psychomotor and mental development is retarded. The karyotype analysis of such an individual will show **PH-II 2022**
(a) Trisomy of chromosome 21
(b) 47 chromosomes with XXY sex chromosomes
(c) 45 chromosomes with XO sex chromosomes
(d) 47 Chromosomes with XYY sex chromosomes

55. Select the incorrect match regarding the symbols used in Pedigree analysis **PH-II 2022**
(a)  – Parent with male child affected with disease
(b)  – Sex unspecified
(c)  – Affected individual
(d)  – Consanguineous mating

56. A normal girl, whose mother is haemophilic marries a male with no ancestral history of haemophilia. What will be the possible phenotypes of the offsprings? **PH-II 2022**
(i) Haemophilic son and haemophilic daughter.
(ii) Haemophilic son and carrier daughter.
(iii) Normal daughter and normal son.
(iv) Normal son and haemophilic daughter. Choose the most appropriate answer from the options given below:
(a) (ii) and (iv) only (b) (i) and (ii) only
(c) (ii) and (iii) only (d) (i) and (iv) only

57. In a cross between a male and female, both heterozygous for sickle cell anaemia gene, what percentage of the progeny will be diseased? **2021**
(a) 100% (b) 50% (c) 75% (d) 25%

58. Select the correct match **2020**

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

59. In which genetic condition, each cell in the affected person, has three sex chromosomes XXY?
(a) Turner's syndrome **2019**
(b) Thalassemia
(c) Klinefelter's syndrome
(d) Phenylketonuria

60. Thalassemia and sickle cell anemia are caused due to a problem in globin molecule synthesis. Select the correct statement: **2017**
(a) Both are due to a quantitative defect in globin chain synthesis
(b) Thalassemia is due to less synthesis of globin molecules
(c) Sickel cell anemia is due to a quantitative problem of globin molecules
(d) Both are due to a qualitative defect in globin chain synthesis

61. A disease caused by an autosomal primary non-disjunction is: **2017**
(a) Klinefelter's Syndrome (b) Turner's Syndrome
(c) Sickel Cell Anemia (d) Down's Syndrome

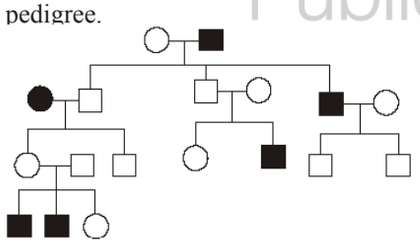
62. Which of the following most appropriately describes haemophilia? **2016**
(a) Recessive gene disorder
(b) X - linked recessive gene disorder
(c) Chromosomal disorder
(d) Dominant gene disorder

63. Pick out the correct statements: **2016**
(i) Haemophilia is a sex-linked recessive disease
(ii) Down's syndrome is due to aneuploidy
(iii) Phenylketonuria is an autosomal recessive gene disorder.
(iv) Sickle cell anaemia is a X-linked recessive gene disorder
(a) (i) and (iv) are correct
(b) (ii) and (iv) are correct
(c) (i), (iii) and (iv) are correct
(d) (i), (ii) and (iii) are correct

64. A cell at telophase stage is observed by a student in a plant brought from the field. He tells his teacher that this cell is not like other cells at telophase stage. There is no formation of cell plate and thus the cell is containing more number of chromosomes as compared to other dividing cells. This would result in **2016**
(a) Aneuploidy (b) Polyploidy
(c) Somaclonal variation (d) Polyteny

65. If a colour-blind man marries a woman who is homozygous for normal colour vision, the probability of their son being colour-blind is **Ph-II 2016**
(a) 0.75 (b) 1 (c) 0 (d) 0.5

66. In the following human pedigree, the filled symbols represent the affected individuals. Identify the type of given pedigree. **2015 RS**



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

67. A colour blind man marries a woman with normal sight who has no history of colour blindness in her family. What is the probability of their grandson being colour blind? **2015 RS**
(a) 1 (b) Nil (c) 0.25 (d) 0.5

68. An abnormal human baby with 'XXX' sex chromosomes was born due to : **2015 RS**
(a) formation of abnormal ova in the mother
(b) fusion of two ova and one sperm
(c) fusion of two sperms and one ovum
(d) formation of abnormal sperms in the father

69. A man whose father was colour blind marries a woman who had a colour blind mother and normal father. What percentage of male children of this couple will be colour blind? **2014**
(a) 25% (b) 0%
(c) 50% (d) 75%

70. A human female with Turner's syndrome: **2014**
(a) has 45 chromosomes with XO.
(b) has one additional X chromosome.
(c) exhibits male characters.
(d) is able to produce children with normal husband.

71. If both parents are carriers for thalassemia, which is an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child? **2013**
(a) 50% (b) 25%
(c) 100% (d) no chance

72. The incorrect statement with regard to Haemophilia is : **2013**
(a) It is a recessive disease
(b) It is a dominant disease
(c) A single protein involved in the clotting of blood is affected
(d) It is a sex-linked disease

73. Which one is the **incorrect** statement with regards to the importance of pedigree analysis **Kar.2013**
(a) It helps to trace the inheritance of a specific trait
(b) It confirms that DNA is the carrier of genetic information
(c) It helps to understand whether the trait in question is dominant or recessive
(d) It confirms that the trait is linked to one of the autosome

74. Down's syndrome in humans is due to **Kar.2013**
(a) Two 'Y' chromosomes
(b) Three 'X' chromosomes
(c) Three copies of chromosome 21
(d) Monosomy

Exercise 6 : Numeral & Event Based MCQs

1. In a certain plant, red fruit (R) is dominant over yellow fruit (r) and tallness (T) is dominant over shortness (t) .If a plant with RRTt genotype is crossed with a plant rrtt genotype , what will be the percentage of tall plants with red fruits in the progeny ?
(a) 50% (b) 100% (c) 75% (d) 25%

2. What is the probability that a cross between a true-breeding pea plant with smooth seeds and a true-breeding pea plant with wrinkled seeds will produce F_1 progeny with smooth seeds ?
(a) 1/2 (b) 1/4 (c) 0 (d) 1

3. Consider a gene that has two alleles and shows complete dominance. When two heterozygotes for this gene breed, they have a 25% chance of producing a homozygous recessive offspring. The next time they breed, what are the chances that they will once again have a homozygous recessive progeny?
(a) 0% (b) 25% (c) 75% (d) 100%

4. Some dogs bark while trailing; others are silent. The barker gene is dominant, the silent gene recessive. These genes are not linked. The gene for normal tail is dominant over the gene for screw (curly) tail. A barker dog with a normal tail that is heterozygous for both traits is mated to another

dog of the same genotype. What fraction of their offspring will be barkers with screw tails?
(a) 3/4 (b) 9/16 (c) 3/16 (d) 1/4

5. There are three genes a, b, c. Percentage of crossing over between a and b is 20%, b and c is 28% an a and c is 8%. What is the sequence of genes on chromosome?
(a) b, a, c (b) a, b, c
(c) a, c, b (d) None of these

6. Red-green colour blindness is a human recessive sex-linked trait. A man and a woman with normal vision have a colour-blind son. What is the probability that their next child will also be a color-blind son?
(a) 0 (b) 1/8 (c) 1/4 (d) 1/2

7. A normal- visioned man whose father was colour. blind, marries a woman whose father was also colour blind. They have their first child as a daughter. what are the chance that this child would be colour blind ?
(a) 100% (b) zero percent
(c) 25% (d) 50%

8. In pea plants, yellow seeds are dominant to green. If a heterozygous yellow seeded plant is crossed with a green seeded plant, what ratio of yellow and green seeded plants would you expect in F_1 generation ?
(a) 9 : 1 (b) 1 : 3 (c) 3 : 1 (d) 50 :50

9. 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) 50% (b) 25% (c) 100% (d) no chance

Hints & Solutions

KEY TERM REFRESHER – FIB

1. Gregor Mendel
2. 14
3. Genes
4. genotype
5. heterozygous
6. factors
7. multiple alleles
8. Sutton and Boveri
9. Drosophila melanogaster
10. Mutation
11. pedigree analysis
12. autosomal recessive trait
13. GAG to GUG
14. aneuploidy
15. 46 (23 pairs)

EXERCISE – 1

1. (d) **2.** (b)

3. (c) Yellow seed colour is dominant over green seed colour.

4. (a) The seven traits are now known to be present on 4 chromosome. But they do not show linkage, because of large distances between them on the chromosome.

5. (a) Black is the phenotype of the sheep.

6. (c) **7.** (a) **8.** (c)

9. (a)

Parents : Tt × tt

Gametes : T t × t

Progeny :

	T	t
t	Tt	tt

Ratio = 1 : 1

10. (a) Test cross is the cross of an individual with an individual having recessive phenotype. It is used to determine the genotype of a plant showing the dominant phenotype, that means to determine whether the individual exhibiting dominating characters are homozygous or heterozygous.

11. (c) Crossing of one F_1 progeny with recessive parent, *e.g.*,

RR (red) × rr (green)

Rr Rr Rr Rr F_1-progeny
Red Red Red Red

F_1-progeny ♀ Rr × ♂ RR

RR Rr Rr RR
red red red red
(pure) (dominant) (dominant) (pure)

To find the genotype of hybrid test cross is done.

12. (c) In order to find out the gamete or the genotype of an unknown individual, Scientists perform a test cross.

13. (c) In test cross, genotype of an organism showing dominant phenotype is determined by crossing it with homozygous recessive genotype.

14. (c) In Co-dominance F_1 generation resemble both the parents. Ex : Blood group inheritance.

15. (b) Inheritance of skin colour in human is controlled by three genes, A, B and C which is polygenic inheritance.

16. (b) Genes coding for the same trait are located at the same locus on homologous chromosomes.

17. (c) The lack of independent assortment in sweet pea and *Drosophila* is due to linkage.

18. (b) Incomplete dominance results in the progeny's expressing an intermediate form of the two parental alleles.

19. (a) The alleles for red and white flowers are co-dominant, resulting in pink flowers when both are present in the genotype.

20. (a) If the mother has blood group I^AI^A and the father has blood group I^BI^O, then their children can only have genotypes I^AI^B or I^AI^O, which have the phenotypes blood group AB and blood group A respectively.

21. (c)

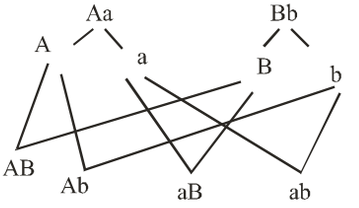
P-generation – RRYY (Round seed yellow cotyledon) × rryy (wrinkled seed green cotyledon)

Gametes : RY ↓ ry

F_1 generation – RrYy (all heterozygous round seeded and yellow cotyledon)

↓ Selfing

22. (d)



So, the corresponding genotype will be AaBb.

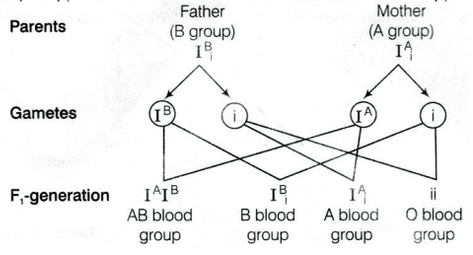
23. (d) Incomplete (partial or mosaic) dominance is the phenomenon where none of the two contrasting alleles or factors is dominant.

24. (a) **25.** (c)

26. **(c)** Pleotropy is a phenomenon in which a single gene may express more than one trait.
27. **(a)**
28. **(b)** In grasshopper the males lack a Y-sex chromosome and have only an X-chromosome. They produce sperm cells that contain either an X chromosome or no sex chromosome, which is designated as O.
29. **(b)** **30.** **(d)**
31. **(c)** Colour blindness in a X-chromosome linked character. So they'll be having all colour blind sons and carrier daughters.
32. **(a)**
33. **(b)** Sickle cell anaemia is caused by a change in a single base pair of DNA.
34. **(a)** Pedigree analysis is a record of the occurrence of a trait in several generations of a human family.
35. **(d)** Mutation can be induced by gamma radiation.
36. **(c)** Type O blood group is considered the universal donor blood group. It can potentially be transfused to any patient regardless of their blood type. In emergency situation, this type of blood can be given to the patient.
37. **(b)** Haemophilia is sex linked recessive disease in which a simple protein that is a part of protein cascade involved in clotting of blood is affected.
38. **(c)** Down's syndrome is the chromosomal disorders due to the presence of an additional copy of the chromosome number 21 (trisomy of 21).

EXERCISE - 2

1. **(b)** **2.** **(d)** **3.** **(b)** **4.** **(a)**
5. **(b)** A cross of F_1 hybrid with its recessive homozygous parent is called the test cross. It is done to determine the genotype of a given plant.
6. **(a)**
7. **(b)** Independent assortment of genes takes place only when they are located on separate non-homologus chromosomes. Where two or more than two genes are located on same chromosome, independent assortment will not be possible.
8. **(b)** "True-breeding" in this case means homozygous for both traits, with the parents of opposite phenotypes.
9. **(b)** The proportion of seeds that turn out to be green and/or wrinkled was very little, indicating that these two are both recessive characters, which are phenotypically expressed only if they are present in homozygous condition.
10. **(d)**
11. **(d)** The mother would have genotype I^AI^B and the father would have genotype I^OI^O.
12. **(c)** When a cross is carried out between heterozygous father (for blood group B) and heterozygous mother (of blood group A) to get four children with different blood groups.



13. **(d)** Linkage is the inheritance of genes of same chromosome together and capacity of these genes to retain their parental combination in subsequent generation.
14. **(d)** ABO blood group system in human beings is an example of codominant, dominant recessive and multiple alletes.
15. **(a)** Tightly linked genes show more linkage then crossing over.
16. **(c)** Polygenic inheritance is the inheritance of traits which are dependent on the no. of genes such as the skin colour of human beings, *eg*. AABB is black AaBB in neither dark nor black. AaBb is again wheatish Aabb is light and aabb is white colour.
17. **(c)** In human the Y chromosome bears the androgenic factor or the male determining factor. Whereas in *Drosophila* the ratio of sex chromosomes to autosomes is the factor determining sex of the individual.
18. **(c)** **19.** **(a)**
20. **(a)** Since the woman's father was colour blind. She would be a carrier of the colour blindness gene. When she marries a colour blind man. Their progeny could be

Parent	X^cX	×	X^cY		
	Carrier Woman		Colour blind Man		
Progeny	X^cX^c	X^cY	XX^c	XY	
	Colour blind son		Carrier daughter		

21. **(d)**
22. **(b)** Pedigree is a chart showing the record of inheritance of certain genetic traits for two or more ancestral generations of an individual, abnormality or disease.
23. **(d)** **24.** **(b)**

EXERCISE - 3

1. **(a)** **2.** **(d)**
3. **(d)** Both the statement are correct.
4. **(d)** Both the statement are correct.
5. **(d)** Both the statement are correct.
6. **(b)**
7. **(b)** Skin colour in humans shows polygenic inheritance.
8. **(d)** In case of incomplete linkage, the linked gene shows new combination along with parental combination due to crossing over between chromatids.
9. **(a)** Aneuploidy can be either due to loss of one or more chromosomes (hypoploidy) or due to addition of one or more chromosomes to complete chromosome complement (hyperploidy).
10. **(b)** In the monohybrid test cross, both dominant and recessive traits are obtained in 1 : 1 ratio.
11. **(d)** F_2 phenotypic and genotypic ratio in monohybrid cross involving incomplete dominance is

1	:	2	:	1
RR		Rr		rr
(red)		(pink)		(white)

12. **(d)** The sex determination in honey bees is based on the number of sets of chromosomes an individual receives.
13. **(d)**
14. **(a)** Statements (i), (iii) and (v) are correct. Mendelian disorders are mainly determined by alternation or mutation in a single gene whereas chromosomal disorders are caused due to absence or excess or abnormal arrangement of one or more chromosomes.
15. **(b)** Linked genes do not undergo hybridisation and thus, they do not affect homozygosity.
16. **(a)** Tightly linked genes show more linkage than crossing over.

17. **(c)** A – IV; B – II; C – I; D – III
18. **(d)** A – II; B – III; C – IV; D – I
19. **(d)** A – III; B – l C – VI, D – V
20. **(c)** A – IV; B – III; C – II; D – I
21. **(c)** A – III; B – I; C – IV; D – II

EXERCISE - 4

1. **(b)** All the genes, present on a particular chromosome form a linkage group.

2. **(a)** The changes in chromosome number by additions or deletions of less than a whole set is aneuploidy. In this case organism gains or loses one or more chromosome but not a complete set.

3. **(a)** Crossing over separates genes aways from each other So, the distance between the genes and percentage of recombination shows and direct relationship, *i.e.*, when genes are close together they have high linkage and exhibit low recombination frequencies.

4. **(d)** Most sex-linked (X-linked) conditions are recessive. This means that person having two X-chromosomes (females), both copies of a gene (*i,e.*, one on each X-chromosome), must have a change or mutation whereas in a person with one X- chromosomes (males), only one copy or a gene must have a mutation.

5. **(d)** Sickle-cell anaemia is an autosome linked recessive trait which is controlled by a single pair of allele Hb^A and Hb^s only the homozygous individuals for Hbs, *i.e.*, Hb^s Hb^s shows the diseased phenotype.

6. **(b)** The example of codominance is ABO blood grouping in humans. ABO blood groups are controlled by gene I.

7. **(d)** In ZZ/ZW case, the female has heteromorphic (ZW) sex chromosomes. Thus peacock shows ZZ/ZW sex determination type.

8. **(b)** The F_1 plants of genotype Tt are self-pollinated. (both tall (T) but with dwarf (t) alleles).
Phenotypic ratio = Tall : Dwarf
3 : 1
Genotypic ratio = Pure tall : Hybrid : Pure dwarf
1 : 2 : 3

9. **(d)** Alleles of two genes are segregeting independently. Mendel explained by crossing a pea plant with round and yellow seeds and one with wrinkled and green ones.
All F_1 hybrids give yellow and round seeds. Since yellow colour is dominant over the green and the round shape is dominant over the winkled.

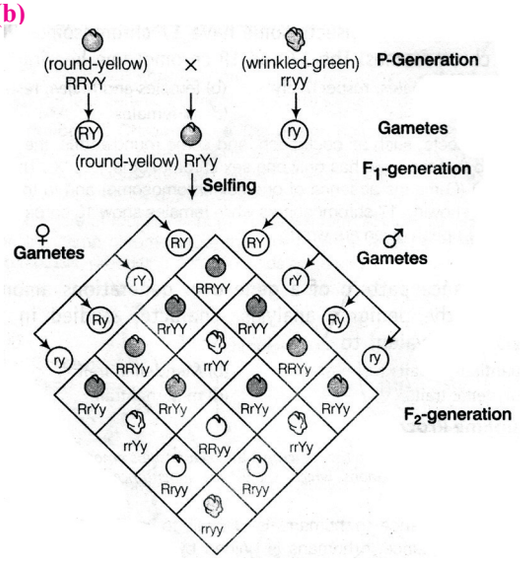
10. **(c)** Linkage will not result in variations among sibling. Morgan carried out several dihybrid crossess in Drosophila to study genes related to sex-linked.
He came to know that the genes were located on the X-chromosome and also observed that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations were much higher than the non-parental type.

11. **(b)** **12.** **(c)**

13. **(a)** In certain insects, such as cockroach, and some roundworms, the Y-chromosome is missing so that the male has only one sex chromosome, *i.e.* 'X' condition in the male is XO (O means absence of one sex chromosome) and in the female it is XX., thus males showing 17 chromosome while females show 18 chromosome.

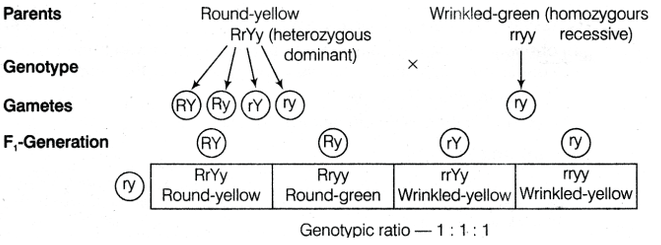
14. **(b)**

15. **(b)**



Phenotypic ratio Round yellow : Round green : Wrinked yellow : Wrinked green
9 : 3 : 3 : 1
Genotypic ratio 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

16. **(a)** 1 : 1 it can be explained by the following test cross.



Genotypic ratio — 1 : 1 : 1 : 1
Genotypic ratio – 1 : 1 : 1 : 1
The other option are incorrect.

17. **(d)** **18.** **(c)**

EXERCISE - 5

1. **(c)** The frequency of recombination is the number of crossovers formed between any two genes. Alfred Sturtevant explained that the recombination frequency between gene pairs is the measure of distance between the genes.

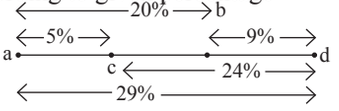
2. **(c)** The phenomenon of pleiotropism refers to phenomenon where a single gene affecting multiple phenotypic expression.

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

6. **(d)** There were 7 characters of the pea plant which were selected by Mendel for the experiments. The characters which were chosen by Mendel for his study were **stem height, flower colour, flower position, pod shape, pod colour, seed shape, seed colour**. All these characters belong to different chromosomes in the pea plant. Therefore both statements are correct.

7. **(b)** Mendel's law of independent assortment states that **the alleles of two (or more) different genes get sorted into gametes independently of one another**.

8. **(c)** According to given percentage



So the sequence of genes, a, b, c & d will be a, c, b, d.

9. **(c)** The chromosomal theory of inheritance was proposed by Sutton and Boveri. 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.

10. **(d)** Since the green pod (G) colour is dominant over yellow pod colour (g) and inflated pod (I) is dominant over constricted pod (i). So,
Green, Inflated Pod X Yellow, constricted Pod
After cross all F_1 progeny will be dominant green, inflated plants and the phenotypic ratio of F_2 plants will be 9:3:3:1.
Green, inflated- 9
Green, constricted- 3
Yellow, inflated- 3
Yellow, constricted- 1
Thus the % of yellow inflated plants will be 3/16= 18.75%.

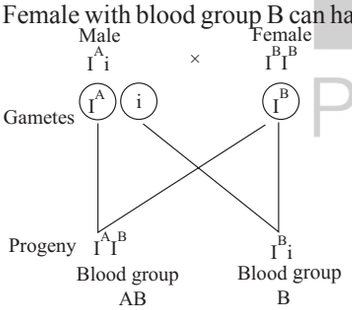
11. **(d)** The production of gametes by the parents, the formation of the zygotes, the F_1 and F_2 plants can be understood from a diagram called punnett square.

12. **(b)** Chromosomal theory of inheritance was proposed by Sutton and Boveri independently in 1902.

13. **(c)** Experimental verification of the chromosomal theory of inheritance was done by Morgan. Sutton and Boveri proposed chromosomal theory of inheritance but it was experimentally verified by T.H. Morgan.

14. **(d)** Punnett square is a graphical representation of the possible genotypes of an offspring arising from a particular cross or breeding event. With the help of Punnett square the production of gametes, the formation of zygotes and the F_1 and F_2 plants can be understood.

15. **(b)** Male with blood group A can have genotype IA i or IAIA.
Female with blood group B can have genotype I^BI^B, or I^Bi



16. **(d)**

17. **(c)** 1 map unit represent 1% cross over. Map unit is used to measure genetic distance. This genetic distance is based on average number of cross over frequency.

18. **(b)** I^AI^O, I^BI^O - Dominant-recessive relationship
I^AI^B - Codominance
I^A, I^B & I^O - Three different allelic forms of a gene (multiple allelism)

19. **(b)** Husband × Wife

♀ \ ♂	I^AI^B	I^Ai
I^A	I^AI^A	I^AI^B
i	I^Ai	I^Bi

Number of genotypes = 4
Number of phenotypes = 3

I^AI^A and I^Ai = A
I^AI^B = AB
I^Bi = B

20. **(a)** During his experiments Mendel have taken seven characters in a pea plant. Among these, nature of trichomes i.e., glandular or non-glandular was not considered by Mendel.

21. **(d)** Mendel conducted hybridisation experiments for 7 years on Pea plant between 1856 to 1863 and his data was published in 1865.

22. **(a)**

23. **(b)** Dominance - (ii) In a heterozygous organism only one allele expresses itself.
Codominance - (iii) In a heterozygous organism both alleles express themselves fully.
Pleiotropy - (iv) A single gene influences many characters.
Polygenic inheritance - (i) Many genes govern a single character.

24. **(c)** When two genes in a dihybrid cross are situated on the same chromosome, the proportion of parental gene combinations are much higher than the non-parental or recombinant type. This is also called **incomplete linkage**.

25. **(a)** Translocation is the mechanism that causes genes to move from one linkage group to another.

26. **(c)** Seven pairs of contrasting characters were selected in pea plant and studied by Mendel in his experiment.

27. **(c)** The most common cause of variations is recombination in the organism which are reproduced by sexual way.

28. **(b)** Alleles are defined as alternative forms of the same gene.

29. **(a)** Mendel did not use pod length for his experiment.

30. **(b)**

Possible genotype of man with blood group A	×	Possible genotype of woman with blood group B
I^AI^A, I^Ai	×	I^BI^B, I^Bi
If the genotype is		
I^Ai	×	I^Bi

The possibility of resultant blood group may be A, B, AB and O.

31. **(b)** All alleles of a gene are situated on the same locus of chromosome in organisms.

32. **(c)** In codominance, both alleles are independently expressed in the heterozygote.

33. **(d)** Morgan observed that while crossing a set of characteristics, two genes did not segregate as per Mendel's law. If two genes were present on the same chromosome, the probability of getting a parental combination was much highr in the next generation as compared to the non-parental combination. He termed this physical association of gene as linkage. So the correct answer is T.H. Morgan.

34. **(c)** A pleiotropic gene regulates multiple traits (characteristics) in an individual.

35. **(b)** Epistasis is the phenomenon of suppression of phenotypic expression of gene by a nonallelic gene which shows its own effect.

36. **(d)** ABO blood group system in human beings is an example of codominant, dominant recessive and multiple alleles. Blood groups are controlled by the gene I located on 9^{th} chromosome that has 3 multiple alleles, out of which any two are found in a person. In codominance, both genes express themselves completely.

37. **(c)** In co-dominance, F_1 generation resembles both the parents. Ex : Blood group inheritance.

38. **(a)** Tightly linked genes show more linkage than crossing over.

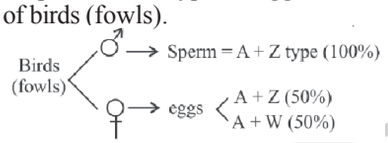
39. **(c)** Crossing over leads to recombination of genetic material on the two chromosomes. Mutation results in alternation of DNA sequences and consequently results in change in the genotype and the phenotype of an organism. In addition to recombination, mutation is another phenomenon that leads to variation in DNA.

40. **(c)**

41. **(b)** XO type of sex-determination is found in grass – hoppers.

42. **(d)** XX- XO type of sex determination is seen in crickets, grasshoppers, and some other insects. In all these organisms the female is XX and is the homogametic sex.

43. **(c)** In birds female heterogamety is found thus sex of progeny depends on the types of egg rather than the type of sperms of birds (fowls).



44. **(c)** Woman acts as a carrier. Both son & daughter inherit X-chromosome. Although only son would be the diseased one.

$$X^cX \times XY$$
$$\downarrow$$
$$X^cX \;\; X^cY \;\; XX \;\; XY$$

45. **(c)** In case of humans, the sex determining mechanism is XY type.

46. **(c)** Statement A, C and D are incorrect w.r.t. Klinefelter's syndrome as they are associated with Down's syndrome.

47. **(a)** Broad palm with single palm crease is visible in a person suffering from Down syndrome.

48. **(b)** Consanguineous marriage is matrimony between individuals who are closely related. In a clinical sense, marriage between two family members who are second cousins or closer qualify as having a consanguineous marriage.
So the correct option is '□=O = mating between relatives'.

49. **(d)** **50.** **(b)**

51. **(a)** Myotonic dystrophy is a autosomal dominant trait.

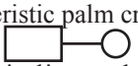
52. **(c)** If female is colour blind → $X^C X^C$ since the mother of man was colour blind, then their will be 100% chance that man will be colour blind C_XC_Y
Hence,

	X^C	Y
X^C	$X^C X^C$	$X^C Y$
X^C	$X^C X^C$	$X^C Y$

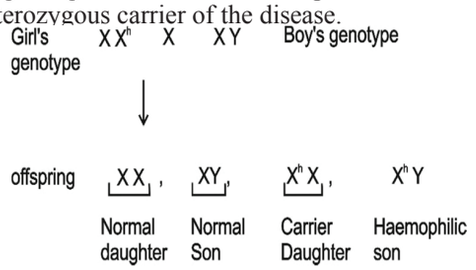
So, all progeny will colour blind.

53. **(a)** In the given statements, Statement I is incorrect and the statement II is correct. The correct statement is Sickle cell anaemia is autosomal recessive disorder and the haemophilia is sex linked recessive disorder.

54. **(a)** If a female individual is with small round head, furrowed tongue, partially open mouth and broad palm with characteristic palm crease.

55. **(d)** Symbol  is used in the human pedigree analysis to indicate the mating between relative (consanguineous mating). So symbol given in the option (iv) is incorrect.

56. **(c)** If a mother is heterozygous (a carrier) for haemophilia and father is normal and not affected by the disorder then each son will have a 1 in 2 (50%) chances of getting his mother's haemophilic allele and get affected by haemophilia. Each daughter has a 1 in 2 (50%) chances of getting her mother's haemophilic allele and being heterozygous carrier of the disease.



57. **(d)** According to given question;

$$Hb^A\ Hb^S \times Hb^A\ Hb^S \quad \text{(Parent)}$$
$$\downarrow$$
$$Hb^S\ Hb^S,\ Hb^A\ Hb^A,\ Hb^S\ Hb^A,\ Hb^S\ Hb^A$$
(Progenies)

Total number of affected progenies = 1
∴ Percentage of diseased/affected progenies
$= \frac{1}{4} \times 100 = 25\%$

58. **(b)** Phenylketonuria is an inborn error of metabolism is also inherited as the autosomal recessive trait. Sickle cell anemia is an autosomes linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous). Thalassemia is an inherited blood disorder that causes your body to have less hemoglobin than normal. Hemoglobin enables red blood cells to carry oxygen. Thalassemia can cause anemia, leaving you fatigued. Haemophilia is a sex linked recessive disease, which shows its transmission from unaffected carrier female to some of the male progeny.

59. **(c)** Klinefelter's syndrome is a chromosomal conditions in boys and men that can affect physical and intellectual development. It is caused due to presence of an additional copy of X-chromosome resulting into 44 + XXY type chromosome complement. Adult with this syndrome may have primary hypogonadism (decreased testosterone production), small or underscent testes, enlarged breast, tall stature etc.

60. **(b)** Thalassemia is a quantitative problem of synthesising very few globin molecules while sickle cell anaemia is a qualitative problem of synthesising an incorrectly functioning globin.

61. **(d)** Down's syndrome is caused by non-disjunction of 21^{st} chromosome i.e. Trisomy.

62. **(b)** Haemophilia A and haemophilia B are inherited in an X-linked recessive pattern. The genes associated with these conditions are located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females

(who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, it is very rare for females to have haemophilia. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

63. **(d)** Sickle cell disease is inherited in an autosomal recessive pattern.

64. **(b)** This phenomenon is known as polyploidy, wherein the cells contain more than two paired (homologous) sets of chromosomes. Polyploidy is often seen in the case of plants. The major cause of polyploidy is the non-disjunction of sister chromatids during meiotic recombination. This condition is actually useful in development of new crop varieties.

65. **(c)** $X^CY \times XX$

(Colours blind man)

then the progeny will be

	X^C	Y
X	X^CX	XY
X	X^CX	XY

$\therefore$ 0% Progeny will be colour blind.

66. **(b)** Autosomal recessive is a type of disorder in which two copies of an abnormal gene must be found for the disease in the affected person.

67. **(c)** $XX \times X^cY$

Normal woman Colourblind man

♀ \ ♂	X^c	Y
X	XX^c	XY
X	XX^c	XY

The daughters of this couple will have normal eye sight but would be carrier. If one of the carrier daughter marries with normal eyed man,

$XX^c \times XY$

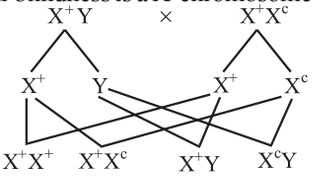
Normal woman Normal man

♀ \ ♂	X	Y
X	XX	XY
X^c	XX^c	X^cY

Only 25% grandson will show colourblindness.

68. **(a)** Triple X syndrome occurs in women, who inherit three X chromosomes--their genotype is XXX. This type of syndrome is very rare and occurs due to defects in meiosis I and II during gamete formation in the mother and produces abnormal ova.

69. **(c)** Colour blindness is a X-chromosome linked character.



$\therefore$ Colourblind male = 50%

70. **(a)** Turner's syndrome is a chromosomal condition that affects development in females. A human female with Turner's syndrome has 45 chromosomes with XO. The most common feature of Turner syndrome is short stature, which becomes evident by about age 5.

71. **(b)** Genotype of carrier parents is – Aa (male parent) × Aa (female parent)

♀ \ ♂	A	a
A	AA	Aa
a	Aa	*aa

AA → normal child (25%)
Aa → carrier child (50%)
aa → affected child (25%)

72. **(b)** Haemophilia is sex linked recessive disease in which a simple protein that is a part of protein cascade involved in clotting of blood is affected. Due to this, in an affected individual, a simple cut will result in non-stop bleeding.

73. **(b)** Pedigree is a chart showing the record of inheritance of certain genetic traits, abnormality or disease for two or more ancestral generations of an individual.

74. **(c)** Down's syndrome is the chromosomal disorder due to the presence of an additional copy of the chromosome number 21 (trisomy of 21). The affected individual is short statured with small round head, furrowed tongue and partially open mouth and mental development is retarted.

EXERCISE - 6

1. **(a)** Parent : RRTt × rrtt

Gametes : (RT), (Rt), (rt)

Offspring : RrTt Rrtt

tall with tall with

Ratio = 1 : 1 red fruit yellow fruit

2. **(d)** This is an example of a monohybrid cross. All of the F_1 progeny would have smooth seeds. (The F_1 generation would all have the genotype Ss, producing the phenotype of smooth seeds because the smooth allele, S, is dominant to the wrinkled allele, s).

3. **(b)** Each conception is an independent event, so the probabilities remain the same each time.

4. **(c)** BbNn × BbNn is expected to produce a ratio of 9 expressing both dominants to 3 expressing one combination of dominant trait and recessive trait to 3 expressing the other combination of dominant and recessive trait to 1 expressing both recessives.

5. **(a)**

6. **(c)** The probability of being male is 1/2. Since they have one color-blind son, the woman must be heterozygous.

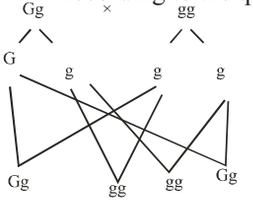
7. **(b)** If a normal visioned man marries a woman whose father was also colourblind. Then his wife would be carried of this disease if her mother was normal. This trait passed in to children but daughters produce by this couple are carrier not the colourblind. 50% of sons would be colourblind.

8. **(d)** Let GG ⇒ homozygous yellow seed plant.

Gg ⇒ heterozygous green

gg ⇒ homozygous green

According to the question :



(yellow) (green) (green) (yellow)

So, the ratio will be 50 : 50

9. **(b)**