1. (a) Define any five of the following: (1x5=5)
    - (i) Missense mutation:
        - A missense mutation is a point mutation in which a single nucleotide change results in a codon that codes for a different amino acid.
        - This can lead to a non-functional protein, a protein with altered function, or no discernible effect.
    - (ii) Monosomy:
        - Monosomy is a form of aneuploidy where there is a missing copy of one chromosome in an otherwise diploid cell.
        - The chromosomal constitution is typically represented as $2n - 1$.
    - (iii) Pure line selection:
        - Pure line selection is a method of plant breeding where a single, homozygous, self-pollinated plant is selected from a mixed population.
        - The progeny of this self-pollinated plant forms a pure line, which is genetically uniform and breeds true.
    - (iv) Pseudodominance:
        - Pseudodominance is the phenotypic expression of a recessive allele when the dominant allele has been deleted.
        - It occurs in individuals heterozygous for a deletion, where the deletion uncovers a recessive allele on the homologous chromosome, making it appear dominant.
    - (v) Lethal alleles:

- Lethal alleles are alleles that cause the death of the organism possessing them, usually during embryonic development or early life.
- They can be dominant, recessive, or conditional, and their presence can alter expected Mendelian ratios.

- (vi) Penetrance:
  - Penetrance is the proportion of individuals in a population who carry a specific genotype and also express the associated phenotype.
  - It describes the probability that a gene will be expressed phenotypically, given that the individual has the gene.

2. (b) Fill in the blanks. Attempt any five (1x5=5)

- (i) The phenomenon in plant breeding where the hybrid offspring exhibits superior traits, such as increased size or yield, compared to both parents is known as **heterosis or hybrid vigor**.
- (ii) In **incomplete or co-dominant** genetic inheritance, neither allele is completely dominant or recessive, and both alleles contribute equally to the phenotype of the offspring.
- (iii) The phenomenon in which a point mutation leads to the premature termination of a polypeptide chain is known as **nonsense** mutation.
- (iv) The experiment by Thomas Hunt Morgan with *Drosophila melanogaster* led to the discovery that the X chromosome carries the gene for red eyes and white eyes, demonstrating the principle of **sex linkage or X-linkage**.
- (v) A chromosomal aberration in which a segment of a chromosome is turned around 180 degrees is called **inversion**.

- (vi) In **sympatric** speciation, speciation occurs typically due to ecological or behavioral factors without physical barriers.

3. (c) Match the following with their contributors (any five): (1x5=5)
    - (i) Chromosomal Theory of Inheritance matched with e. Theodore Boveri & Walter Sutton
    - (ii) Plant Selection Method in plant breeding matched with d. Nilsson-Ehle
    - (iii) Linkage matched with f. Thomas Hunt Morgan
    - (iv) Coining of the term 'Genetics' matched with a. William Bateson
    - (v) DNA structure matched with b. Watson and Crick
    - (vi) Genetic Mapping matched with c. Alfred Sturtevant
4. Differentiate between any five of the following: (3x5=15)
    - (i) Complete linkage and incomplete linkage
        - **Complete Linkage:**
            - Genes are located very close together on the same chromosome and are always inherited together.
            - No crossing over occurs between them.
            - Only parental combinations are observed in the progeny.
        - **Incomplete Linkage:**
            - Genes are located on the same chromosome but are far enough apart for crossing over to occur between them.

- Both parental and recombinant combinations are observed in the progeny, with parental combinations being more frequent.
- The frequency of recombination is proportional to the distance between the genes.

- (ii) Test cross and reciprocal cross
  - **Test Cross:**
    - A cross between an individual of unknown genotype (usually dominant phenotype) and a homozygous recessive individual.
    - Used to determine the genotype of the unknown individual.
    - If the unknown is heterozygous, recessive phenotypes will appear in the progeny.
  - **Reciprocal Cross:**
    - Two crosses in which the genotypes of the parents are reversed with respect to sex.
    - For example, if in the first cross, female A is crossed with male B, in the reciprocal cross, female B is crossed with male A.
    - Used to determine if a trait is sex-linked or maternally inherited.
- (iii) Polyploidy and aneuploidy
  - **Polyploidy:**
    - A chromosomal abnormality where an organism has more than two complete sets of chromosomes.
    - Examples include triploidy ($3n$), tetraploidy ($4n$), etc.

- Often results in larger cells and organisms and can be beneficial in plant breeding.

▪ **Aneuploidy:**

- A chromosomal abnormality where there is an abnormal number of chromosomes within a set (e.g., one or more chromosomes are added or deleted).
- Examples include monosomy ($2n - 1$), trisomy ($2n + 1$), etc.
- Often leads to developmental abnormalities and is generally detrimental to the organism.

○ (iv) Point mutations and frame shift mutations

▪ **Point Mutations:**

- Involve a change in a single nucleotide base within the DNA sequence.
- Can be silent, missense, or nonsense mutations.
- Do not alter the reading frame of the gene.

▪ **Frameshift Mutations:**

- Involve the insertion or deletion of nucleotides in a DNA sequence, in numbers not divisible by three.
- Alter the reading frame of the gene, leading to a completely different amino acid sequence downstream of the mutation.
- Often result in non-functional proteins due to premature stop codons or extensive amino acid changes.

○ (v) Pure line selection and Mass selection

- **Pure Line Selection:**
    - Selection within a population of self-pollinated plants, starting from a single, homozygous plant.
    - Progeny are genetically identical to the parent (except for new mutations).
    - Aims to develop genetically uniform varieties.
- **Mass Selection:**
    - Selection of individual plants based on their phenotype from a mixed population, and the seeds from these selected plants are bulked to constitute the next generation.
    - The population remains genetically heterogeneous.
    - Aims to improve the average performance of the population.

- (vi) Maternal inheritance and maternal effect
    - **Maternal Inheritance:**
        - The phenotype of the offspring is determined by the genotype of the mother's cytoplasm, specifically by genes located in organelles like mitochondria or chloroplasts.
        - The paternal genes have no influence on the trait.
        - Inheritance pattern does not follow Mendelian ratios.
    - **Maternal Effect:**
        - The phenotype of the offspring is determined by the genotype of the mother, but the genes are nuclear (not cytoplasmic).

- Maternal gene products (e.g., proteins or RNA) deposited in the egg determine the early developmental stages or traits of the offspring.
- The offspring's own genotype for the trait does not manifest until later in development.

5. Write short notes on any five of the following: (3x5=15)
    - (i) Dosage compensation
        - Dosage compensation is a genetic mechanism that equalizes the expression of genes located on the sex chromosomes between males and females.
        - In mammals, it involves the inactivation of one of the two X chromosomes in females, forming a Barr body. This ensures that females, with two X chromosomes, produce the same amount of X-linked gene products as males, who have only one X chromosome.
        - In *Drosophila*, dosage compensation involves hypertranscription of the single X chromosome in males.
        - This mechanism is crucial for normal development and survival, as an imbalance in X-linked gene products can be lethal.
    - (ii) Mitochondrial inheritance
        - Mitochondrial inheritance is a form of non-Mendelian inheritance where traits are passed down exclusively from the mother to all her offspring.
        - Mitochondria contain their own circular DNA (mtDNA) and are typically inherited cytoplasmically through the egg cell, as sperm contributes very few or no mitochondria to the zygote.

- Diseases caused by mutations in mtDNA, such as Leber's Hereditary Optic Neuropathy (LHON), are examples of mitochondrial inheritance, affecting all children of an affected mother but none of the children of an affected father.

- (iii) Interspecific hybridization
  - Interspecific hybridization is the process of crossing individuals from two different species to produce a hybrid offspring.
  - This can occur naturally or through artificial means in plant and animal breeding.
  - The goal is often to combine desirable traits from both parent species or to create new genetic variations.
  - Hybrid offspring are often sterile (e.g., mule, a hybrid of a horse and a donkey) due to differences in chromosome number or structure, which prevents proper meiosis.
  - In plants, interspecific hybridization can be used to transfer disease resistance or other valuable traits from wild species to cultivated crops.
- (iv) Transposable elements
  - Transposable elements (TEs), also known as "jumping genes," are DNA sequences that can move from one location in the genome to another.
  - They were discovered by Barbara McClintock in maize.
  - There are two main classes: Class 1 (retrotransposons) move via an RNA intermediate, and Class 2 (DNA transposons) move directly as DNA.

- TEs can cause mutations by inserting into genes, altering gene expression, or promoting chromosomal rearrangements.
- They play a significant role in genome evolution and can be a source of genetic variation.

- (v) Chemical mutagens
  - Chemical mutagens are chemical agents that can cause changes in the DNA sequence, leading to mutations.
  - They work through various mechanisms, including base modification (e.g., alkylating agents, deaminating agents), base analogs (molecules structurally similar to normal bases that can be incorporated into DNA), and intercalating agents (molecules that insert themselves between DNA base pairs).
  - Examples include nitrous acid (deamination), ethyl methanesulfonate (EMS) (alkylation), and acridine dyes (intercalation).
  - Exposure to chemical mutagens can increase the risk of genetic diseases and cancer.
- (vi) Down's syndrome
  - Down's syndrome, also known as Trisomy 21, is a chromosomal disorder caused by the presence of an extra copy of chromosome 21.
  - It is the most common chromosomal disorder in humans, characterized by a distinct set of physical features, intellectual disability, and increased risk of certain health problems (e.g., heart defects, gastrointestinal abnormalities).

- The extra chromosome 21 is usually a result of non-disjunction during meiosis in either the egg or sperm cell.
- The incidence of Down's syndrome increases with maternal age.

6. (a) In a fruit fly, the genes for body color (B=black, b=grey) and wing size (W=long, w=vestigial) are linked. A test cross was done between a heterozygous black body, long winged fly (BbWw) and a homozygous recessive gray body, vestigial winged fly (bbww). Of the 200 offsprings produced, 180 had parental phenotypes (black body, long wings or gray body, vestigial wings) and 20 had the recombinant phenotypes (black body, vestigial wings or gray body, long wings). What is the recombination frequency between these two genes? (6)
    - Total number of offspring = 200
    - Number of parental phenotypes = 180
    - Number of recombinant phenotypes = 20
    - Recombination frequency is calculated as:
      Recombination frequency $= \frac{\text{Number of recombinant offspring}}{\text{Total number of offspring}} \times 100\%$
    - Recombination frequency = $(20/200) \times 100\%$
    - Recombination frequency = $0.1 \times 100\%$
    - Recombination frequency = 10%
    - The recombination frequency between the genes for body color and wing size is 10%.
7. (b) What is epistasis? Explain the types of epistasis with examples. (3+6=15)
    - **Epistasis:**

- Epistasis is a phenomenon where the expression of one gene (epistatic gene) masks or modifies the effect of another gene (hypostatic gene) at a different locus.
- It results in deviations from the expected Mendelian dihybrid cross ratios (e.g., 9:3:3:1), as the interaction between genes affects the phenotype.

- **Types of Epistasis with Examples:**
  - **1. Recessive Epistasis (Supplementary Epistasis):**
    - One gene, when homozygous recessive, masks the expression of another gene at a different locus.
    - Typical F2 ratio: 9:3:4.
    - **Example: Coat color in Labrador Retrievers.**
      - Gene E/e controls pigment deposition: EE or Ee allows pigment deposition, ee prevents pigment deposition (yellow).
      - Gene B/b controls pigment color: BB or Bb produces black, bb produces brown.
      - An individual with genotype ee will be yellow, regardless of the B/b genotype, because the ee genotype prevents pigment from being deposited at all.
      - If E_B_ = black, E_bb = brown, eeB_ = yellow, eebb = yellow.
  - **2. Dominant Epistasis:**
    - A dominant allele at one locus masks the expression of alleles at a second locus.
    - Typical F2 ratio: 12:3:1.

- **Example: Fruit color in Summer Squash.**
    - Gene W/w: W (dominant white) is epistatic to other color genes, w (recessive colored).
    - Gene Y/y: Y (dominant yellow), y (recessive green).
    - If W_Y_ = white, W_yy = white.
    - Only when homozygous recessive for W (ww) can the color gene express: wwY_ = yellow, wwyy = green.

- **3. Duplicate Recessive Epistasis (Complementary Genes):**
    - When either of two genes is homozygous recessive, it masks the expression of the dominant allele at the other locus. Both dominant alleles are required together for a particular phenotype.
    - Typical F2 ratio: 9:7.
    - **Example: Flower color in Sweet Peas.**
        - Two genes, C and P, are required to produce purple pigment.
        - C_P_ = purple; C_pp = white; ccP_ = white; ccpp = white.
        - If an individual is homozygous recessive for either gene (cc or pp), the purple pigment cannot be formed, resulting in a white flower.
- **4. Duplicate Dominant Epistasis (Redundant Genes):**
    - The presence of a dominant allele at either of two loci produces the same phenotype. Only the

homozygous recessive genotype at both loci results in a different phenotype.

- Typical F2 ratio: 15:1.
- **Example: Fruit shape in Shepherd's Purse.**
    - Two genes, A and B, determine triangular fruit shape.
    - A_B_ = triangular; A_bb = triangular; aaB_ = triangular.
    - Only aabb = ovoid (non-triangular).

- **5. Inhibitory Epistasis (Dominant and Recessive Interaction):**
    - A dominant allele at one locus inhibits the expression of a dominant allele at a second locus.
    - Typical F2 ratio: 13:3.
    - **Example: Feather color in Chickens.**
        - Gene I/i: I (dominant inhibitor) suppresses color; i (recessive non-inhibitor).
        - Gene C/c: C (dominant color); c (recessive white).
        - I_C_ = white; I_cc = white.
        - Only when homozygous recessive for I (ii) can the color gene express: iiC_ = colored; iicc = white.

8. (a) Explain the inheritance of kappa particles in Paramecium. (7)
    - The inheritance of kappa particles in *Paramecium* is a classic example of cytoplasmic or non-Mendelian inheritance,

specifically demonstrating a symbiotic relationship with a genetic basis.

- **Kappa particles** are bacterial endosymbionts (*Caedibacter taeniospiralis*) found in the cytoplasm of some strains of *Paramecium aurelia*, known as "killer" strains.
- These killer strains produce a toxic substance, called **paramecin**, which is lethal to other "sensitive" strains of *Paramecium* that lack kappa particles.
- **Genetic Control:** The maintenance and replication of kappa particles are dependent on a dominant nuclear gene, **K**, in the *Paramecium* host.
    - *Paramecium* cells with genotype K_K_ or K_k_ can maintain kappa particles.
    - *Paramecium* cells with genotype k_k_ cannot maintain kappa particles, even if they initially receive them.
- **Inheritance Pattern:**
    - When a killer strain (K_K_ with kappa) mates with a sensitive strain (k_k_ without kappa) through conjugation, the inheritance of kappa particles is primarily cytoplasmic.
    - **Brief Conjugation:** If conjugation is brief, there is usually no cytoplasmic exchange. The F1 progeny will have K_k_ genotype, but only the progeny from the killer parent will have kappa particles and remain killer. The progeny from the sensitive parent will be sensitive, as they did not receive kappa.
    - **Prolonged Conjugation:** If conjugation is prolonged, there is cytoplasmic exchange between the two conjugants. In this case, the sensitive conjugant may receive kappa particles from the killer conjugant.

- After prolonged conjugation, both conjugants will have the K_k_ genotype in their nucleus.
- Both conjugants will also have kappa particles in their cytoplasm and will give rise to killer progeny.
- However, if these K_k_ killer progeny are subsequently grown under conditions that cause the loss of kappa particles (e.g., high temperature, starvation), and then allowed to self-fertilize, the resulting k_k_ individuals (from Mendelian segregation) will be unable to regain or maintain kappa particles, even if exposed to them.

- **Conclusion:** The presence of kappa particles is necessary for the killer phenotype, but their persistence is governed by the dominant nuclear gene K in the *Paramecium* host. This illustrates the interplay between nuclear genes and cytoplasmic factors in determining a phenotype.

9. (b) Explain chloroplast and mitochondrial inheritance with suitable examples. (4+4=8)

- **Chloroplast Inheritance:**
  - **Explanation:** Chloroplasts are organelles found in plant cells that contain their own circular DNA (cpDNA) and are responsible for photosynthesis. Chloroplast inheritance refers to the transmission of genetic traits exclusively through the chloroplast genome. In most flowering plants, chloroplasts are inherited maternally, meaning they are passed down only from the egg cell, while the pollen contributes virtually no chloroplasts to the zygote.
  - **Key Characteristics:**
    - Uniparental inheritance (typically maternal).
    - Does not follow Mendelian ratios.

- All offspring from a cross will exhibit the chloroplast-determined phenotype of the maternal parent.
- Reciprocal crosses yield different results.

- **Suitable Example: Variegation in *Mirabilis jalapa* (Four o'clock plant).**
  - The *Mirabilis jalapa* plant exhibits three types of branches/leaves: green, white, and variegated (patches of green and white).
  - This trait is determined by the chloroplasts.
  - If flowers from a green branch are pollinated:
    - By pollen from a green branch → All offspring are green.
    - By pollen from a white branch → All offspring are green.
    - By pollen from a variegated branch → All offspring are green.
  - If flowers from a white branch are pollinated:
    - By pollen from any type of branch → All offspring are white (and often die young due to lack of chlorophyll).
  - If flowers from a variegated branch are pollinated:
    - By pollen from any type of branch → Offspring can be green, white, or variegated, depending on which type of ovule (containing green, white, or mixed chloroplasts) is fertilized.
  - This maternal inheritance pattern clearly shows that the phenotype is determined by the chloroplasts

present in the egg cell, not by the nuclear genes from the pollen.

- **Mitochondrial Inheritance:**
  - **Explanation:** Mitochondria are organelles found in eukaryotic cells that contain their own circular DNA (mtDNA) and are primarily responsible for cellular respiration. Mitochondrial inheritance refers to the transmission of genetic traits exclusively through the mitochondrial genome. In most sexually reproducing organisms, mitochondria are inherited maternally, as the egg cell contributes the vast majority of mitochondria to the zygote, while the sperm contributes very few or none.
  - **Key Characteristics:**
    - Uniparental inheritance (typically maternal).
    - Does not follow Mendelian ratios.
    - All offspring of an affected mother will inherit the trait.
    - None of the offspring of an affected father will inherit the trait (unless there's rare paternal leakage).
  - **Suitable Example: Leber's Hereditary Optic Neuropathy (LHON) in Humans.**
    - LHON is a genetic disorder that causes rapid, painless loss of vision due to the degeneration of the optic nerve.
    - It is caused by mutations in mitochondrial DNA (mtDNA).
    - **Inheritance Pattern:**

- If a mother has LHON (and thus, carries the mutated mtDNA), all of her children (sons and daughters) will inherit the mutated mitochondria and are at risk of developing the disease.
- If a father has LHON, none of his children will inherit the mutated mitochondria or the disease, because he does not pass on his mitochondria to his offspring.

- This pattern directly reflects the maternal inheritance of mitochondria.

10. (a) In a population of 1,000 individuals, 360 individuals have blue eyes (recessive trait). What are the allele frequencies for blue eyes (b) and brown eyes (B)? (7)

- Given:
  - Total population size (N) = 1,000 individuals
  - Number of individuals with blue eyes (recessive trait) = 360
- Let:
  - 'b' represent the allele for blue eyes (recessive)
  - 'B' represent the allele for brown eyes (dominant)
- Individuals with blue eyes have the genotype 'bb'.
- Frequency of homozygous recessive genotype (q^2) = (Number of blue-eyed individuals) / (Total population size)
- $q^2 = 360/1000 = 0.36$
- To find the frequency of the recessive allele 'b' (q), take the square root of q^2:

- $q = \sqrt{0.36} = 0.6$
- So, the allele frequency for blue eyes (b) is 0.6.
- According to the Hardy-Weinberg principle, $p + q = 1$, where 'p' is the frequency of the dominant allele (B) and 'q' is the frequency of the recessive allele (b).
- $p = 1 - q$
- $p = 1 - 0.6 = 0.4$
- So, the allele frequency for brown eyes (B) is 0.4.
- **Answer:**
  - Allele frequency for blue eyes (b) = 0.6
  - Allele frequency for brown eyes (B) = 0.4

11. (b) What is Barr body? How is it a consequence of dosage compensation? Explain different genetic mechanisms of sex determination in living organisms.

- **Barr Body:**
  - A Barr body is an inactivated X chromosome found in the somatic cells of most female mammals.
  - It appears as a dense, darkly staining structure in the interphase nucleus.
  - It is named after Murray Barr, who first observed it.
  - The formation of a Barr body is a random process, meaning that in different cells of the same female, either the maternal or paternal X chromosome can be inactivated. This leads to mosaicism in females for X-linked traits.
- **How it is a consequence of dosage compensation:**

- Dosage compensation is a genetic mechanism that ensures equal expression of X-linked genes in individuals with different numbers of X chromosomes (e.g., males XY and females XX).
- In mammals, this is achieved through **X-inactivation**, where one of the two X chromosomes in female somatic cells is largely silenced.
- The Barr body is the physical manifestation of this epigenetically silenced, condensed X chromosome.
- By inactivating one X chromosome, female cells effectively have only one active X chromosome, similar to males. This balances the "dosage" of X-linked gene products between the sexes, preventing an overdose of these gene products in females and ensuring proper development and function.

- **Different Genetic Mechanisms of Sex Determination in Living Organisms:**
  - **1. XY Sex Determination (e.g., Humans, Mammals, *Drosophila*):**
    - Males are heterogametic (XY) and produce two types of gametes (X and Y).
    - Females are homogametic (XX) and produce only one type of gamete (X).
    - In humans, the presence of the SRY (Sex-determining Region Y) gene on the Y chromosome triggers male development.
    - In *Drosophila*, the ratio of X chromosomes to autosomes determines sex (X:A ratio). An X:A ratio of 1.0 (XX) is female, and 0.5 (XY or XO) is male.

- **2. ZW Sex Determination (e.g., Birds, Butterflies, Moths, some Reptiles and Fish):**
    - Females are heterogametic (ZW) and determine the sex of the offspring.
    - Males are homogametic (ZZ).
    - The Z chromosome carries genes important for male development, and the W chromosome is usually smaller and contains fewer genes, often associated with female determination.
- **3. XO Sex Determination (e.g., Grasshoppers, Crickets, many Insects):**
    - Males have only one sex chromosome (XO), lacking a second sex chromosome. They are heterogametic.
    - Females have two X chromosomes (XX) and are homogametic.
    - Sex is determined by the number of X chromosomes. An even number of X chromosomes (XX) results in a female, while an odd number (XO) results in a male.
- **4. Haplodiploidy (e.g., Bees, Ants, Wasps - Hymenoptera):**
    - Sex is determined by the number of chromosome sets.
    - Fertilized eggs develop into diploid (2n) females (queens and workers).
    - Unfertilized eggs develop into haploid (n) males (drones).

- Males have only one set of chromosomes, while females have two sets.

- **5. Environmental Sex Determination (ESD) (e.g., some Reptiles like Crocodiles, Turtles, some Fish):**
    - Sex is not determined by genetic factors (chromosomes) but by environmental cues, most commonly temperature during a critical period of embryonic development.
    - **Temperature-Dependent Sex Determination (TSD):**
        - In some species, specific incubation temperatures lead to male development, while others lead to female development.
        - For example, in many turtles, cooler temperatures produce males, and warmer temperatures produce females (F-M pattern).
        - In some crocodiles, intermediate temperatures produce males, while very high or very low temperatures produce females (F-M-F pattern).
- **6. Single Gene Sex Determination (e.g., some Fungi, some Algae, some Plants):**
    - In some organisms, sex is determined by alleles at a single gene locus, rather than by distinct sex chromosomes.
    - Different alleles at this locus can lead to the development of different sexes or mating types.

These diverse mechanisms highlight the evolutionary flexibility in how sex is determined across different life forms.