1. (a) Distinguish between the following (Any three): (2×3=6)

(i) Co-dominance and Incomplete dominance

- **Co-dominance:**
    - Both alleles in a heterozygous individual are fully expressed, resulting in offspring with a phenotype that is a combination of both parental phenotypes, not an intermediate.
    - Examples include human ABO blood groups where both A and B alleles are expressed in AB blood type.
- **Incomplete dominance:**
    - Neither allele is completely dominant over the other, leading to a heterozygous phenotype that is intermediate between the two homozygous phenotypes.
    - Examples include the snapdragon flower color, where a cross between red and white flowers produces pink flowers.



(ii) Coupling and Repulsion

- **Coupling (cis arrangement):**
    - Dominant alleles of two different genes are located on the same homologous chromosome, and their recessive alleles are located on the other homologous chromosome.
    - For example, if gene A and B are linked, then AB/ab represents coupling.
- **Repulsion (trans arrangement):**
    - The dominant allele of one gene is on one homologous chromosome, and the recessive allele of the other gene is on the same chromosome, while the other homologous chromosome carries the recessive allele of the first gene and the dominant allele of the second gene.

- For example, if gene A and B are linked, then Ab/aB represents repulsion.

(iii) Sex-influenced and sex-limited inheritance

- **Sex-influenced inheritance:**
  - The expression of a trait is influenced by the sex of the individual, but the trait itself is not carried on a sex chromosome. The same genotype can produce different phenotypes in males and females.
  - Examples include pattern baldness in humans, where the allele for baldness acts as dominant in males but recessive in females.
- **Sex-limited inheritance:**
  - A trait is expressed in only one sex, even if both sexes have the same genotype. The expression is completely limited to one sex.
  - Examples include milk production in cows or plumage patterns in some bird species, where certain traits are only expressed in females or males, respectively.



(iv) Conservative and replicative transposons

- **Conservative transposons (cut-and-paste transposons):**
  - These transposons are excised from their original location in the genome and inserted into a new location.
  - The number of copies of the transposon in the genome does not increase.
  - This mechanism involves DNA intermediates.
- **Replicative transposons (copy-and-paste transposons):**
  - These transposons make a copy of themselves, and this copy is then inserted into a new location in the genome, while the original copy remains at its original site.

- The number of copies of the transposon in the genome increases.
- This mechanism can involve either DNA or RNA intermediates (retrotransposons).

(b) Define the following (Any three): (1×3=3)

(i) Multiple alleles

- Multiple alleles refer to the existence of more than two alternative forms of a gene that can occupy the same locus on a chromosome. While an individual can only possess two alleles for a given gene (one from each parent), a population can have many different alleles for that gene. An example is the ABO blood group system in humans, which is determined by three alleles ($I^A$, $I^B$, and i).

(ii) Episome

- An episome is a plasmid that can exist either as an independent, self-replicating extrachromosomal DNA molecule or can integrate reversibly into the host cell's chromosome. Examples include the F factor (fertility factor) in *E. coli* which can exist freely in the cytoplasm or integrate into the bacterial chromosome.



(iii) Heterosis

- Heterosis, also known as hybrid vigor, is the phenomenon where the progeny resulting from a cross between two genetically distinct parents (often inbred lines) show superior performance (e.g., increased growth rate, yield, fertility, or resistance to disease) compared to the average of their parents. This is often attributed to the masking of deleterious recessive alleles and/or the synergistic interaction of favorable dominant alleles in the heterozygous state.

(iv) Pleiotropy

- Pleiotropy is a phenomenon where a single gene affects multiple, seemingly unrelated phenotypic traits. Instead of a one-to-one relationship between a gene and a trait, one gene product can have diverse effects on different

physiological processes or structures. An example is the gene responsible for phenylketonuria (PKU) in humans, which, when mutated, can lead to intellectual disability, light skin and hair, and other symptoms.

(c) Give the significant contribution of the following scientists (Any three): (1×3=3)

(i) Alfred Sturtevant

- Alfred Sturtevant, a student of Thomas Hunt Morgan, is credited with constructing the first genetic linkage map (chromosome map) using recombination frequencies. His work demonstrated that the frequency of crossing over between genes could be used to determine their relative distances on a chromosome.

(ii) William Bateson

- William Bateson was a prominent English biologist who coined the term "genetics" to describe the study of heredity and variation. He was a strong advocate for Mendelian principles and conducted extensive research on inheritance patterns, including discovering genetic linkage and coining terms like "allelomorphs" (later shortened to alleles) and "epistasis."



(iii) C B Bridges

- Calvin Bridges, another prominent member of Thomas Hunt Morgan's "fly room," provided strong evidence for the chromosome theory of inheritance. He demonstrated that non-disjunction of sex chromosomes in *Drosophila* led to predictable patterns of abnormal sex chromosome numbers and associated phenotypes, thus directly linking genes to chromosomes.

(iv) Barbara McClintock

- Barbara McClintock was an American geneticist who discovered transposable elements (also known as "jumping genes") in maize. Her groundbreaking work showed that certain genetic elements could move from one position to another within the genome, influencing gene expression and causing mutations. She was awarded the Nobel Prize in Physiology or Medicine in 1983 for this discovery.

(d) Give reasons for the following (Any two): (1.5×2=3)

(i) Some XX humans were found to be males and XY humans were found to be females.

- This phenomenon can be explained by the presence or absence of the SRY gene (Sex-determining Region Y gene).
- **XX males:** These individuals have a translocation of the SRY gene from the Y chromosome onto one of their X chromosomes during paternal meiosis. Even though they have an XX genotype, the presence of the SRY gene on the X chromosome triggers the development of male characteristics.
- **XY females:** These individuals typically have a deletion or mutation in the SRY gene on their Y chromosome, or a defect in the receptors for male hormones. Without a functional SRY gene product, or the ability to respond to androgens, the default female developmental pathway proceeds, leading to an XY genotype with a female phenotype.



(ii) A cross between pure line sinistrally-coiled shell female Limnaea and dextrally coiled shell male Limnaea yielded all sinistrally-coiled shell progeny.

- This is an example of **maternal effect inheritance**.
- In *Limnaea* (a freshwater snail), the direction of shell coiling (dextral or sinistral) is determined by the genotype of the mother, not the genotype of the individual snail itself.
- The mother's genotype determines the substances deposited in the egg cytoplasm, which in turn controls the early developmental stages that dictate the coiling direction.
- Since the female parent in this cross was a pure line sinistrally-coiled shell female, all her progeny will inherit the cytoplasmic factors that lead to sinistral coiling, regardless of the male's genotype.

(iii) For a paracentric inversion, with rare exceptions, recombinant chromosomes are not stable and will not lead to viable offspring.

- A **paracentric inversion** is an inversion that does not include the centromere.
- When a heterozygote for a paracentric inversion undergoes meiosis, a loop forms during synapsis to allow homologous regions to pair.
- If a single crossover occurs within this inversion loop, it leads to the formation of two types of abnormal recombinant chromatids:
    - One **dicentric chromatid** (having two centromeres). This chromatid will be pulled to opposite poles during anaphase I, often breaking due to tension, leading to acentric fragments or bridges.
    - One **acentric chromatid** (lacking a centromere). This chromatid will be lost during cell division as it cannot attach to the spindle fibers.
- The resulting gametes from such a meiotic event will be deficient in some genes and duplicated in others, or will contain broken chromosomes. Such gametes are generally non-viable, leading to zygotic lethality or inviable offspring. The only viable offspring typically arise from non-recombinant chromatids.



2. (a) What do you understand by gene interactions? Discuss any two types of gene interactions (with suitable examples) that cause deviation from the Mendel's dihybrid ratio. (10)

- **Gene interactions** refer to the way different genes or alleles at different loci interact with each other to influence a single phenotype. While Mendel's dihybrid crosses often showed a 9:3:3:1 phenotypic ratio for two independently assorting genes, many complex traits involve interactions between genes, leading to modifications or deviations from this classical ratio. These interactions can occur at various levels, from the biochemical pathways controlled by gene products to the developmental processes they regulate.
- **Two types of gene interactions causing deviation from Mendel's dihybrid ratio:**

- **i) Epistasis:**
  - **Definition:** Epistasis is a form of gene interaction where one gene (the epistatic gene) masks or modifies the expression of another gene (the hypostatic gene) at a different locus. This is not about one allele masking another at the same locus (dominance), but rather one gene influencing the expression of another gene.
  - **Mechanism:** Epistasis often occurs when genes in a biochemical pathway interact sequentially. For example, one gene might encode an enzyme necessary for the production of a precursor molecule, and if this gene is mutated to a non-functional form, the downstream gene, even if functional, cannot produce its product.
  - **Example: Recessive Epistasis (9:3:4 ratio)**
    - **Context:** Coat color in Labrador retrievers.
    - **Genes Involved:** Two genes, B/b (black/brown pigment) and E/e (pigment deposition).
      - B (black pigment), b (brown pigment)
      - E (allows pigment deposition in hair), e (prevents pigment deposition, leading to yellow color regardless of B/b)
    - **Cross:** A dihybrid cross between two BbEe individuals.
    - **Genotypes and Phenotypes:**
      - B_E_: Black (e.g., BBEE, BBEe, BbEE, BbEe)
      - bbE_: Chocolate/Brown (e.g., bbEE, bbEe)
      - B_ee: Yellow (e.g., BBee, Bbee)
      - bbee: Yellow

- **Explanation:** The "ee" genotype is epistatic to the B/b gene. If a dog is "ee," no pigment will be deposited in the hair, resulting in a yellow phenotype, irrespective of whether the B/b gene is for black or brown pigment. This means B_ee and bbee genotypes both result in yellow dogs.
- **Resulting Ratio:** A cross between two dihybrids (BbEe x BbEe) yields a phenotypic ratio of **9 Black : 3 Chocolate : 4 Yellow**, which deviates from the Mendelian 9:3:3:1 ratio.

- **ii) Complementary Gene Interaction (9:7 ratio):**
  - **Definition:** Complementary gene interaction occurs when two dominant alleles from two different genes are required together for the expression of a particular phenotype. If either one or both dominant alleles are absent, a different (often recessive) phenotype is expressed. This implies that the products of both genes are necessary for the completion of a pathway.
  - **Mechanism:** This usually happens when two different genes control sequential steps in a biochemical pathway, and both steps are required to produce the final product that results in the observed phenotype.
  - **Example: Flower color in sweet peas (*Lathyrus odoratus*)**
    - **Context:** Purple vs. White flower color.
    - **Genes Involved:** Two genes, C/c and P/p.
      - Gene C produces enzyme C, which converts a colorless precursor into an intermediate colorless compound.
      - Gene P produces enzyme P, which converts the intermediate colorless compound into the purple pigment.

- **Cross:** A dihybrid cross between two CcPp individuals.
- **Genotypes and Phenotypes:**
    - C_P_: Purple (e.g., CCPP, CcPP, CCPp, CcPp) - Both dominant alleles are present, allowing the pathway to complete.
    - C_pp: White (e.g., CCpp, Ccpp) - Enzyme P is absent, so the intermediate cannot be converted to purple pigment.
    - ccP_: White (e.g., ccPP, ccPp) - Enzyme C is absent, so the precursor cannot be converted to the intermediate.
    - ccpp: White - Both enzymes are absent.
- **Explanation:** For purple color to appear, both the dominant allele C and the dominant allele P must be present. If either is homozygous recessive (cc or pp), the flowers will be white.
- **Resulting Ratio:** A cross between two dihybrids (CcPp x CcPp) yields a phenotypic ratio of **9 Purple : 7 White**, a clear deviation from the 9:3:3:1 Mendelian ratio.



3. (b) Explain the sex-linked inheritance with any one example. (5)

- **Sex-linked inheritance** refers to the inheritance of genes located on the sex chromosomes (X or Y chromosomes in humans, Z or W in birds). Since males and females have different complements of sex chromosomes, the inheritance patterns of these genes differ significantly from those of autosomal genes. In humans, most sex-linked traits are X-linked, meaning the genes are on the X chromosome, as the Y chromosome carries very few genes.
- **Key features of X-linked recessive inheritance (most common type):**

- Males (XY) are hemizygous for X-linked genes, meaning they have only one copy of each gene on the X chromosome. Therefore, any allele present on their single X chromosome will be expressed, regardless of whether it is dominant or recessive.
- Females (XX) have two copies of each X-linked gene. They can be homozygous dominant, heterozygous, or homozygous recessive. Recessive traits are only expressed if they are homozygous recessive.
- Affected males cannot pass the trait to their sons (as they pass their Y chromosome to sons), but they pass their X chromosome to all their daughters, making all their daughters carriers if the mother is not affected.
- Affected females pass the trait to all their sons.
- The trait often appears more frequently in males than in females.
- There is no father-to-son transmission of the trait.



- **Example: Red-Green Color Blindness in Humans**
  - **Gene Location:** The genes responsible for red and green color vision are located on the X chromosome.
  - **Alleles:**
    - $X^C$: Normal color vision allele (dominant)
    - $X^c$: Color blindness allele (recessive)
  - **Genotypes and Phenotypes:**
    - **Females:**
      - $X^CX^C$: Normal vision
      - $X^CX^c$: Normal vision (carrier)
      - $X^cX^c$: Color blind
    - **Males:**

- $X^CY$: Normal vision
- $X^cY$: Color blind

- **Inheritance Pattern Example:**
  - Consider a cross between a normal vision father ($X^CY$) and a carrier mother ($X^CX^c$).
  - **Father's Gametes:** $X^C$, Y
  - **Mother's Gametes:** $X^C$, $X^c$
  - **Possible Offspring Genotypes and Phenotypes:**
    - $X^CX^C$: Daughter with normal vision
    - $X^CX^c$: Daughter with normal vision (carrier)
    - $X^CY$: Son with normal vision
    - $X^cY$: Son who is color blind
  - **Observations:**
    - All daughters will have normal vision (50% will be carriers).
    - 50% of sons will have normal vision, and 50% of sons will be color blind.
    - This clearly illustrates how the trait appears more frequently in males and how females can be carriers without expressing the trait.

3. (a) A female Drosophila heterozygous at three loci- cu/cu+ (curved vs. straight wings), e/e+ (ebony vs. gray bodies), st/st+ (scarlet vs. red eyes) was test crossed with completely homozygous recessive males. The following progeny were observed: cu e st+ = 366 cu+ e+ st = 380 cu e st = 24 cu+ e+ st+ = 30 cu+ e st = 89 cu e+ st+ = 105 cu e+ st = 2 cu+ e st+ = 4

(i) Are the three genes linked? Give reason for your answer.

- **Reasoning:** Yes, the three genes are linked.
- In a test cross involving independently assorting genes, the expected phenotypic ratio of the progeny would be 1:1:1:1:1:1:1:1. This means all eight possible combinations of alleles (parental and recombinant) would be present in approximately equal proportions.
- However, in the observed progeny data, the parental combinations (cu e st+ = 366 and cu+ e+ st = 380) are significantly more frequent than the recombinant combinations. The recombinant classes, especially the double crossover classes (cu e+ st = 2 and cu+ e st+ = 4), are much rarer. This unequal distribution, with a strong prevalence of parental types and much lower frequencies of recombinant types, is a clear indication that the genes are linked and located on the same chromosome.

(ii) What is the order of genes?



- To determine the order of genes, we need to identify the double crossover (DCO) classes. DCO classes are the least frequent recombinant classes.
- Parental classes: cu e st+ (366) and cu+ e+ st (380)
- The DCO classes are cu e+ st (2) and cu+ e st+ (4).
- Compare the DCO classes with the parental classes. In a DCO, the middle gene's alleles are "swapped" relative to the parental arrangement.
- Parental arrangement: cu e st+ and cu+ e+ st
- DCO arrangement: cu e+ st and cu+ e st+
- Observing the DCOs, we see that the allele 'e+' (from cu+ e+ st) has swapped its position relative to 'st' and 'cu', becoming 'cu e+ st'. Similarly, 'e' (from cu e st+) has swapped, becoming 'cu+ e st+'.
- Therefore, **e (ebony/gray body) is the middle gene.**
- The order of genes is **cu - e - st**.

(iii) Determine the map distance and construct a linkage map.

- Total progeny = 366 + 380 + 24 + 30 + 89 + 105 + 2 + 4 = 1000
- **Calculate recombination frequency between cu and e (RFcu-e):**
    - This involves single crossovers between cu and e, and double crossovers.
    - Single crossovers between cu and e: (cu+ e st) = 89, (cu e+ st+) = 105
    - Double crossovers: (cu e+ st) = 2, (cu+ e st+) = 4
    - RFcu-e = (Number of SCO between cu and e + Number of DCO) / Total progeny * 100
    - RFcu-e = (89 + 105 + 2 + 4) / 1000 * 100
    - RFcu-e = (200) / 1000 * 100 = 20%
    - Map distance between cu and e = **20 cM**
- **Calculate recombination frequency between e and st (RFe-st):**
    - This involves single crossovers between e and st, and double crossovers.
    - Single crossovers between e and st: (cu e st) = 24, (cu+ e+ st+) = 30
    - Double crossovers: (cu e+ st) = 2, (cu+ e st+) = 4
    - RFe-st = (Number of SCO between e and st + Number of DCO) / Total progeny * 100
    - RFe-st = (24 + 30 + 2 + 4) / 1000 * 100
    - RFe-st = (60) / 1000 * 100 = 6%
    - Map distance between e and st = **6 cM**
- **Construct a linkage map:**

  cu -------- 20 cM -------- e -------- 6 cM -------- st

(iv) Calculate the coefficient of coincidence and interference.

- **Coefficient of Coincidence (C):**
  - C = Observed number of DCO / Expected number of DCO
  - Observed DCO = 2 + 4 = 6
  - Expected DCO = RFcu-e * RFe-st * Total progeny
  - Expected DCO = 0.20 * 0.06 * 1000
  - Expected DCO = 0.012 * 1000 = 12
  - C = 6 / 12 = **0.5**
- **Interference (I):**
  - I = 1 - C
  - I = 1 - 0.5 = **0.5**
  - This means there is 50% interference, indicating that the occurrence of one crossover reduces the probability of a second crossover in the adjacent region by 50%.



4. (b) Explain the cytological basis of crossing over with the help of an experiment. (6)

- **Cytological Basis of Crossing Over:**
  - Crossing over is the physical exchange of genetic material between homologous chromosomes during prophase I of meiosis. This process results in the recombination of alleles and contributes to genetic variation.
  - **Mechanism:**
    - **Synapsis:** During zygotene of prophase I, homologous chromosomes pair up precisely, forming a bivalent (or tetrad). This pairing is facilitated by the synaptonemal complex.

- **Chiasma Formation:** In pachytene, segments of non-sister chromatids of homologous chromosomes break at corresponding points. These broken ends then rejoin, but with the non-sister chromatid, forming a cross-shaped structure called a chiasma (plural: chiasmata). Each chiasma represents a point where crossing over has occurred.
- **Recombinant Chromatids:** As meiosis proceeds through diplotene and diakinesis, the homologous chromosomes separate, but remain connected at the chiasmata. By the end of meiosis I, each homologous chromosome still consists of two sister chromatids, but now these chromatids are a mix of parental and recombinant segments.
- **Separation:** In meiosis II, sister chromatids finally separate, leading to gametes that contain unique combinations of alleles.



- **Experiment demonstrating the Cytological Basis of Crossing Over (Creighton and McClintock's experiment with Maize, or Stern's experiment with Drosophila):**
  - We will explain the experiment conducted by Harriet Creighton and Barbara McClintock in **maize** (1931), which provided direct cytological evidence that genetic recombination (crossing over) is accompanied by a physical exchange of chromosome segments.
  - **Objective:** To demonstrate that genetic crossing over is indeed a physical exchange between homologous chromosomes.
  - **Experimental Setup:**
    - They used two specific homologous chromosomes in maize that were cytologically distinguishable (i.e., they had visible morphological markers) and also carried different genetic markers (alleles).
    - **Chromosome 9:** They focused on chromosome 9, which in a particular maize strain had two distinct morphological features:

- One homolog had a heavily stained knob at one end.
- The other homolog had a translocated piece of chromosome 8 at the other end (making it longer).

- **Genetic Markers:** On this same chromosome 9, they identified two genes with known alleles:
  - **Colored (C) vs. Colorless (c) kernels:** C is dominant to c.
  - **Waxy (Wx) vs. Starchy (wx) endosperm:** Wx is dominant to wx.
- **Parental Cross:** They crossed a heterozygous plant that was **genetically heterozygous** (C wx / c Wx) and **cytologically heterozygous** (one chromosome had the knob and the C allele, and was homozygous recessive for waxy, while the other homologous chromosome had the translocation and the c allele, and was homozygous dominant for waxy).
  - Parental Genotype/Cytotype: (Knob C wx -- Translocation) / (No Knob c Wx -- No Translocation)
  - Effectively: (Knob-C-wx-Long) and (NoKnob-c-Wx-Short)
- They performed a test cross by crossing this dihybrid parent with a homozygous recessive plant (c wx / c wx) which had morphologically normal (non-knobbed, non-translocated) chromosome 9s.

- **Predictions:**
  - If genetic recombination (crossing over) involved a physical exchange of chromosomal segments, then recombinant progeny (e.g., those with c wx or C Wx phenotypes) should show a corresponding change in their chromosome morphology (e.g., a knobbed chromosome also having the Wx allele, or a non-

knobbed chromosome having the wx allele and the translocation).

- If crossing over was merely a "switching" of genetic information without physical exchange, then the chromosomal morphology should remain the same as the original parental chromosomes.

- **Results and Conclusion:**

  - Creighton and McClintock analyzed the phenotypes of the offspring and then, crucially, examined the cytology of their chromosome 9s.

  - They found a direct correlation: whenever genetic recombination occurred (producing recombinant phenotypes like "colorless and waxy" (c wx) or "colored and starchy" (C Wx)), the chromosomes of these offspring also showed novel combinations of the cytological markers (knob and translocation).



  - For example, if an offspring showed the "c wx" phenotype (recombinant), its chromosome 9 was found to have the "no-knob" end but also the "translocation" end, indicating that a physical exchange had occurred between the knobbed and non-knobbed homologous chromosomes.

  - This direct correspondence between genetic recombination and the exchange of cytological markers provided conclusive evidence that **crossing over involves the physical breakage and reciprocal exchange of segments between homologous chromosomes**.

4. (a) Discuss the molecular basis of spontaneous and induced mutations. Differentiate between aneuploidy and polyploidy with suitable examples. (5+4=9)

- **Molecular Basis of Spontaneous Mutations:**

- Spontaneous mutations are naturally occurring changes in the DNA sequence that arise from errors in cellular processes or interactions with the cellular environment, without any exposure to external mutagens.
- **Molecular Mechanisms:**
  - **Tautomeric Shifts:** DNA bases can exist in alternative tautomeric forms (e.g., keto-enol for thymine and guanine, amino-imino for adenine and cytosine). These rare forms can alter base-pairing properties (e.g., enol-G pairs with T, imino-A pairs with C), leading to misincorporation of nucleotides during DNA replication and subsequent point mutations (transitions: purine-to-purine or pyrimidine-to-pyrimidine substitutions; transversions: purine-to-pyrimidine or pyrimidine-to-purine substitutions).
  - **Slippage during Replication (Indels):** During DNA replication, especially in regions with repetitive sequences (e.g., trinucleotide repeats), the DNA polymerase can "slip," causing either the addition (insertion) or deletion of nucleotides. This can lead to frameshift mutations if the number of inserted or deleted bases is not a multiple of three.
  - **Depurination/Depyrimidination:** The N-glycosidic bond linking a purine (A or G) or pyrimidine (C or T) base to the deoxyribose sugar can spontaneously hydrolyze, leading to the loss of the base and formation of an apurinic (AP) or apyrimidinic site. If not repaired, DNA polymerase may insert a random base opposite the AP site, often leading to a transversion mutation.
  - **Deamination:** Spontaneous deamination of cytosine results in uracil, which pairs with adenine instead of guanine. If not repaired before replication, this leads to a C-G to T-A transition. Similarly, deamination of adenine yields

hypoxanthine, which pairs with cytosine instead of thymine, leading to an A-T to G-C transition.

- **Oxidative Damage:** Reactive oxygen species (ROS) produced during normal metabolism can damage DNA bases, leading to various lesions (e.g., 8-oxoguanine, which pairs with adenine instead of cytosine), causing G-C to T-A transversions.

- **Molecular Basis of Induced Mutations:**
  - Induced mutations are caused by external agents called mutagens, which can be physical or chemical agents that alter the DNA sequence.
  - **Molecular Mechanisms:**
    - **Alkylating Agents (e.g., Ethylmethane Sulfonate - EMS):** These chemicals add alkyl groups to DNA bases. For example, EMS can alkylate guanine, causing it to mispair with thymine, leading to G-C to A-T transitions.
    - **Intercalating Agents (e.g., Ethidium Bromide, Acridine Dyes):** These flat, planar molecules insert themselves between adjacent base pairs in the DNA helix. This distortion can lead to insertions or deletions of base pairs during DNA replication, resulting in frameshift mutations.
    - **Base Analogs (e.g., 5-Bromouracil - 5-BU):** These are chemicals that are structurally similar to normal DNA bases and can be incorporated into DNA during replication. For instance, 5-BU is an analog of thymine but can exist in a rare enol form that pairs with guanine, leading to A-T to G-C transitions.
    - **Ionizing Radiation (e.g., X-rays, Gamma rays):** These high-energy radiations cause ionization of molecules, leading to the formation of highly reactive free radicals. These radicals can cause various types of DNA damage, including single-strand and double-strand breaks, deletions, translocations, and various

base modifications. Double-strand breaks are particularly dangerous as they can lead to large chromosomal rearrangements.

- **UV Radiation (e.g., UV-C):** UV radiation, particularly at 260 nm, is absorbed by pyrimidines, causing the formation of pyrimidine dimers (thymine dimers or cytosine dimers). These dimers distort the DNA helix and block DNA replication and transcription, often leading to errors during repair or replication, resulting in point mutations.

- **Differentiate between Aneuploidy and Polyploidy:**
  - **Aneuploidy:**
    - **Definition:** Aneuploidy refers to a condition where an organism has an abnormal number of chromosomes, involving the gain or loss of one or more individual chromosomes, but not a complete set.
    - **Mechanism:** It typically arises from non-disjunction during meiosis (failure of homologous chromosomes or sister chromatids to separate properly), leading to gametes with n-1 or n+1 chromosomes.
    - **Types:**
      - **Monosomy (2n-1):** Loss of one chromosome from a diploid set.
        - *Example:* Turner syndrome in humans (45, X), where an individual has only one X chromosome instead of two.
      - **Trisomy (2n+1):** Gain of one chromosome to a diploid set.
        - *Example:* Down syndrome in humans (Trisomy 21), where there are three copies of chromosome

21 instead of two. Klinefelter syndrome (47, XXY) and Patau syndrome (Trisomy 13) are other examples.

- **Nullisomy (2n-2):** Loss of both homologous chromosomes of a pair.
- **Tetrasomy (2n+2):** Gain of two homologous chromosomes of a pair.

- **Effect:** Aneuploidy generally has severe phenotypic consequences and is often lethal in animals due to gene dosage imbalance. In humans, it is a major cause of miscarriages and genetic disorders.

- **Polyploidy:**
  - **Definition:** Polyploidy is a condition where an organism has more than two complete sets of chromosomes. The entire genome is duplicated multiple times.
  - **Mechanism:** It arises from various events, such as errors in meiosis leading to diploid gametes, fertilization between unreduced gametes, or replication errors in somatic cells that are not followed by cell division. It can also be induced artificially.
  - **Types:**
    - **Autopolyploidy:** All sets of chromosomes originate from the same species (e.g., triploid, tetraploid).
      - *Example:* Triploid watermelons (3n) are sterile and seedless. Many cultivated plants like potatoes (tetraploid) are autopolyploids.
    - **Allopolyploidy:** Sets of chromosomes originate from two or more different species, usually through hybridization followed by chromosome doubling.

- *Example:* Wheat (*Triticum aestivum*) is a hexaploid (6n) resulting from ancient hybridization events between three different diploid species.

- **Effect:** Polyploidy is often lethal in animals (especially vertebrates) but is common and often advantageous in plants, leading to increased vigor, larger size, and adaptability. It plays a significant role in plant evolution and crop breeding.

5. (b) Describe the Ac-Ds elements in maize. Comment on the significance of P elements. (4+2=6)

- **Ac-Ds elements in Maize (Activator-Dissociation):**
  - The Ac and Ds elements are the first transposable elements discovered by Barbara McClintock in maize, leading to her Nobel Prize. They are Class II DNA transposons, meaning they move via a "cut-and-paste" mechanism, involving a DNA intermediate.
  - **Ac (Activator) element:**
    - The Ac element is an **autonomous** transposable element. This means it contains the gene that encodes for the enzyme **transposase**, which is necessary for its own transposition (movement) as well as the transposition of Ds elements.
    - Ac elements are typically around 4.5 kb in length and have inverted terminal repeats (ITRs) at their ends, which are recognized by the transposase enzyme.
  - **Ds (Dissociation) element:**
    - The Ds element is a **non-autonomous** transposable element. This means it lacks the functional transposase gene and therefore cannot transpose on its own.
    - Ds elements are often shorter than Ac elements and are typically derived from Ac elements through deletions or other mutations within the transposase gene.

- For a Ds element to transpose, an active Ac element (which provides the transposase enzyme) must be present elsewhere in the genome.

- **Mechanism of Action and Phenotypic Effects:**
  - McClintock observed that the presence and movement of these elements caused unstable mutations, leading to variegated (mottled or spotted) phenotypes, particularly in maize kernel color.
  - For example, if a Ds element inserts into a gene responsible for pigment production (e.g., a gene for purple color), it disrupts the gene, leading to a colorless phenotype.
  - However, if an Ac element is also present, it can cause the Ds element to excise from the gene during somatic development. When Ds excises, the gene's function can be restored, leading to patches of pigmented tissue (e.g., purple spots on a colorless background).
  - The timing and frequency of Ds excision, controlled by Ac, determine the size and number of these pigmented spots, resulting in the characteristic variegation.



- **Significance of P elements:**
  - **P elements** are another type of Class II DNA transposon found in *Drosophila melanogaster*. They are structurally and functionally similar to Ac-Ds elements, with autonomous P elements encoding a transposase and non-autonomous P elements (missing parts of the transposase gene) requiring an autonomous P element for their movement.
  - **Significance:**
    - **Hybrid Dysgenesis:** P elements are famous for causing a syndrome called hybrid dysgenesis in *Drosophila*. This occurs when males carrying P elements (P strains) are crossed with

females lacking them (M strains). The offspring (hybrids) exhibit high rates of sterility, mutations, and chromosome rearrangements due to uncontrolled P element transposition in the germline. This phenomenon is critical for understanding the regulation of transposon activity.

- **Tool in Genetic Engineering and Research:** P elements have been extensively exploited as powerful tools in *Drosophila* genetics and molecular biology.
    - **Germline Transformation:** Modified P elements are used as vectors to introduce foreign genes into the *Drosophila* germline, allowing for the creation of transgenic flies. This is a fundamental technique for studying gene function, gene expression, and creating disease models.
    - **Mutagenesis:** P elements can be used to generate insertional mutations by randomly inserting into genes, thereby disrupting their function. This allows researchers to identify and characterize new genes involved in various biological processes.
    - **Gene Trapping:** P elements can be engineered to carry reporter genes (e.g., GFP) that are only expressed when the P element inserts into an active gene, allowing for the identification and visualization of expressed genes.
    - **Enhancer Trapping:** They can also be designed to identify regulatory elements (enhancers) by integrating near genes and driving reporter gene expression in specific patterns.
- In summary, P elements, primarily due to their role in hybrid dysgenesis and their manipulability, have revolutionized *Drosophila* genetic research, making *Drosophila* a leading

model organism for understanding gene function and development.

5. (a) Compare the mechanisms of dosage compensation in humans and Drosophila. How many Ban bodies will be observed in the individuals with Klinefelter syndrome and with Patau syndrome? (7+2=9)

- **Comparison of Dosage Compensation in Humans and Drosophila:**

| Feature | Humans (Mammals) | Drosophila |
|---|---|---|
| **Sex Chromosomes** | XX (female), XY (male) | XX (female), XY (male) |
| **Mechanism** | **X-inactivation (Lyonization)** | **Hypertranscription of the single X chromosome in males** |
| **Target Sex** | Females (XX) | Males (XY) |
| **Process** | One of the two X chromosomes in XX individuals is largely inactivated and condensed into a Barr body. This occurs randomly in early embryonic development in somatic cells. | The single X chromosome in XY males is transcribed at approximately twice the rate of a single X chromosome in XX females. |
| **Gene Expression Level** | Each active X chromosome expresses X-linked genes at roughly the same level as the single X in males. The inactive X is largely silenced. | The genes on the single X chromosome in males produce a similar amount of product as the genes on both active X chromosomes in females (each producing half the amount). |

| Feature | Humans (Mammals) | Drosophila |
|---|---|---|
| **Molecular Basis** | Involves the *XIST* (X-inactive specific transcript) gene, which produces a long non-coding RNA that coats and silences the inactive X chromosome. Leads to heterochromatin formation. | Involves a complex called the Male-Specific Lethal (MSL) complex, which binds to specific sites on the male X chromosome and acetylates histones, leading to a more open chromatin structure and increased transcription. |
| **Visible Feature** | Formation of a **Barr body** (condensed, inactive X chromosome) in the nuclei of somatic cells of XX individuals. The number of Barr bodies is typically N-1, where N is the number of X chromosomes. | No visible condensed structure like a Barr body. The compensation is achieved through transcriptional upregulation. |
| **Mosaicism** | Females are **mosaics** for X-linked traits because the random inactivation of either the paternal or maternal X chromosome in different cell lineages leads to patches of cells expressing different alleles. | No mosaicism for X-linked traits based on dosage compensation; expression is uniform across cells. |
| **Evolutionary Origin** | Thought to have evolved to balance gene dosage between XX and XY individuals. | Thought to have evolved to balance gene dosage between XX and XY individuals. |

- **Number of Barr bodies in individuals with Klinefelter syndrome and Patau syndrome:**
    - **Barr body formula:** Number of Barr bodies = (Total number of X chromosomes) - 1
    - **Klinefelter syndrome:**
        - Individuals with Klinefelter syndrome typically have a karyotype of **47, XXY**.
        - Total number of X chromosomes = 2
        - Number of Barr bodies = 2 - 1 = **1 Barr body**
    - **Patau syndrome:**
        - Individuals with Patau syndrome have **Trisomy 13**, meaning they have an extra copy of chromosome 13. Their sex chromosomes are typically normal, either XX or XY.
        - If the individual is **47, XX, +13**:
            - Total number of X chromosomes = 2
            - Number of Barr bodies = 2 - 1 = **1 Barr body**
        - If the individual is **47, XY, +13**:
            - Total number of X chromosomes = 1
            - Number of Barr bodies = 1 - 1 = **0 Barr bodies**
        - Therefore, in an individual with Patau syndrome, the number of Barr bodies will depend on their sex chromosome complement: **1 if female (XX), 0 if male (XY)**.

5. (b) Compare the phenomena of nuclear and extranuclear inheritance. Explain the inheritance of pigmentation in Ephestia. (3+3=6)

- **Comparison of Nuclear and Extranuclear Inheritance:**

| Feature | Nuclear Inheritance (Mendelian Inheritance) | Extranuclear Inheritance (Cytoplasmic/Mitochondrial/Chloroplast Inheritance) |
|---|---|---|
| **Genetic Material** | DNA located in the nucleus, organized into chromosomes. | DNA located in organelles like mitochondria and chloroplasts (and sometimes plasmids/viruses). |
| **Location of Genes** | Genes are located on nuclear chromosomes. | Genes are located in the cytoplasm, within specific organelles. |
| **Transmission** | Inherited from both parents (biparental) through gametes (sperm and egg contribute nuclear DNA equally). | Typically inherited predominantly or exclusively from one parent, usually the mother (maternal inheritance), via the cytoplasm of the egg. Paternal contribution of organelles is rare or absent. |
| **Mendelian Ratios** | Follows Mendelian laws of segregation and independent assortment, resulting in predictable phenotypic | Does not follow Mendelian ratios. Reciprocal crosses often yield different results. |

| Feature | Nuclear Inheritance (Mendelian Inheritance) | Extranuclear Inheritance (Cytoplasmic/Mitochondrial/Chloroplast Inheritance) |
|---|---|---|
| | ratios (e.g., 3:1, 9:3:3:1). | |
| **Reciprocal Crosses** | Yield similar results (e.g., A x B is same as B x A for autosomal traits). | Often yield different results, as the maternal parent largely determines the phenotype of the offspring. |
| **Effect of Sex** | Sex can influence expression (sex-linked, sex-influenced, sex-limited) but not the fundamental inheritance pattern of nuclear genes. | Primarily maternal inheritance (e.g., mitochondrial diseases are passed from mother to all children). |
| **Examples** | Blood groups, eye color, height in humans; pea plant traits (color, shape). | Mitochondrial diseases, variegation in plants, kappa particles in *Paramecium*, male sterility in corn. |



- **Inheritance of Pigmentation in *Ephestia* (Flour Moth):**

- The inheritance of pigmentation (larval color) in the flour moth *Ephestia kuehniella* is a classic example of **maternal effect inheritance**, a type of extranuclear inheritance. In this case, the phenotype of the offspring is determined by the genotype of the mother, specifically by substances deposited in the egg cytoplasm during oogenesis, rather than the offspring's own genotype.
- **Background:**
  - Larvae can have pigmented (dark) skin or unpigmented (reddish-brown) skin.
  - The pigmentation is controlled by a single gene with two alleles:
    - **A (dominant):** Produces kynurenine, a precursor for pigment synthesis.
    - **a (recessive):** Does not produce kynurenine.
  - Individuals with the "A" allele produce an enzyme that converts tryptophan into kynurenine, leading to pigmentation. Individuals with "aa" genotype cannot produce kynurenine.
- **The Maternal Effect:**
  - If the mother has at least one dominant "A" allele (i.e., her genotype is AA or Aa), she will produce kynurenine. This kynurenine is then deposited into the cytoplasm of all the eggs she produces.
  - Even if an offspring inherits the "aa" genotype from its parents, as long as its mother was AA or Aa, it will initially develop as a pigmented larva because it utilizes the kynurenine (and the enzyme that processes it) pre-supplied by the mother's cytoplasm.
  - As the larva develops, the maternally supplied kynurenine (or enzyme) is eventually used up. At this point, the larva's own

genotype takes over. If its genotype is "aa," it will then lose pigmentation and become unpigmented. However, the initial larval color is determined by the mother.

- **Cross Example:**
  - **Cross 1: Pigmented Female (Aa) x Unpigmented Male (aa)**
    - Female (Aa) produces eggs, all of which contain maternally deposited kynurenine (because she has the 'A' allele).
    - Offspring genotypes: 50% Aa, 50% aa
    - **Phenotype of all larvae:** All larvae will initially be **pigmented**, regardless of their own genotype. This is because all eggs received kynurenine from the mother's cytoplasm.
  - **Cross 2: Unpigmented Female (aa) x Pigmented Male (AA or Aa)**
    - Female (aa) produces eggs that **do not** contain maternally deposited kynurenine (because she lacks the 'A' allele).
    - Offspring genotypes: All will be Aa (if male was AA) or 50% Aa, 50% aa (if male was Aa).
    - **Phenotype of all larvae:** All larvae will be **unpigmented**, regardless of their own genotype, because their mother (aa) did not deposit kynurenine in the eggs. Even if they inherited the dominant 'A' allele from the father, the lack of initial maternal supply prevents early pigmentation.
- **Conclusion:** This demonstrates that the larval pigmentation in *Ephestia* is a maternal effect trait. The phenotype of the offspring's larval stage is determined by the genotype of the mother, specifically by the presence or absence of the kynurenine precursor in the egg

cytoplasm, rather than the direct expression of the offspring's own genes at that early stage.

6. Write short notes on any three of the following: (3×5=15)

(a) CIB method of detection of mutations

- The **CIB method** is a classical genetic technique developed by Hermann Muller in *Drosophila melanogaster* to efficiently detect recessive lethal mutations on the X chromosome. It was instrumental in demonstrating that X-rays could induce mutations, for which Muller later received the Nobel Prize.
- **CIB stands for:**
    - **C:** Crossover suppressor (an inversion on the X chromosome that prevents crossing over with its homolog, ensuring the entire X chromosome is inherited as a block).
    - **l:** A recessive lethal allele already present on the CIB chromosome. This allele ensures that any male inheriting the CIB chromosome will die, and any female homozygous for the CIB chromosome will also die.
    - **B:** Bar eyes, a dominant visible marker on the CIB chromosome, which allows for easy tracking of the CIB chromosome.
- **Methodology:**
    a. **Generation of Mutated X Chromosomes:** Males are exposed to a mutagen (e.g., X-rays) to induce mutations, particularly on their X chromosome.
    b. **Mating 1:** These mutagenized males are crossed with virgin CIB females.
        - CIB females are heterozygous for the CIB chromosome and a normal X chromosome ($X^{C}IB\ X^{+}$).

- The F1 generation will include female offspring that are heterozygous for the CIB chromosome and a mutagenized X chromosome ($X^{C}IB\ X^{mut}$). These females have Bar eyes due to the 'B' allele on the CIB chromosome. Males inheriting the CIB chromosome die (due to 'l'), and males inheriting the mutagenized X chromosome survive.

c. **Mating 2:** Individual F1 Bar-eyed females ($X^{C}IB\ X^{mut}$) are then mated with wild-type males ($X^{+}$ Y).

d. **Observation of F2 Progeny:**

- In the F2 generation, males inheriting the CIB chromosome ($X^{C}IB$ Y) will die due to the 'l' allele.
- Males inheriting the $X^{mut}$ chromosome ($X^{mut}$ Y) are observed.
- **If the $X^{mut}$ chromosome carries a newly induced recessive lethal mutation:** All males receiving this chromosome will die. Therefore, the F2 culture bottle will contain **no male offspring** (only females).
- **If the $X^{mut}$ chromosome does not carry a new lethal mutation:** Both males ($X^{mut}$ Y) and females will be present in the F2 generation.

- **Significance:** The CIB method allows for the rapid and efficient screening of a large number of X chromosomes for newly induced recessive lethal mutations in a single generation of F2 males. It provides a powerful tool for studying mutagenesis and identifying genes essential for viability.

(b) Retrotransposons

- **Retrotransposons**, also known as Class I transposable elements, are a type of transposable element that move within the genome via an **RNA intermediate**. Unlike DNA transposons (Class II), which use a "cut-and-paste" or "copy-and-paste" DNA mechanism, retrotransposons utilize a "copy-and-paste" mechanism involving reverse transcription. They are

abundant in eukaryotic genomes, often comprising a significant portion of the genome (e.g., over 40% in humans).

- **Mechanism of Transposition:**

    e. **Transcription:** The retrotransposon DNA sequence within the genome is first transcribed into an RNA molecule.

    f. **Reverse Transcription:** This RNA molecule then serves as a template for the enzyme **reverse transcriptase** (which is either encoded by the retrotransposon itself or provided by another retrotransposon/virus). Reverse transcriptase synthesizes a complementary DNA (cDNA) copy from the RNA template.

    g. **Integration:** The newly synthesized cDNA copy is then inserted into a new location in the host cell's genome, typically by an enzyme called **integrase** (also often encoded by the retrotransposon).

- **Key Characteristics:**

    - **Increase in Copy Number:** Since the original retrotransposon remains at its location and a new copy is inserted elsewhere, retrotransposon transposition leads to an increase in the number of copies of the element in the genome over generations.

    - **Structural Features:** Many retrotransposons resemble retroviruses in their structure, particularly those with long terminal repeats (LTRs).

    - **Types:**

        - **LTR Retrotransposons:** These elements have long terminal repeats (sequences of several hundred base pairs) at their ends. They encode genes for reverse transcriptase and integrase (and sometimes other genes similar to retroviral genes, like gag and pol). Examples include Ty elements in yeast and copia elements in *Drosophila*.

        - **Non-LTR Retrotransposons:** These elements lack LTRs. They typically have a poly-A tail at one end. They include:

- **LINEs (Long Interspersed Nuclear Elements):** Autonomous retrotransposons that encode reverse transcriptase and an endonuclease. They are relatively long (e.g., L1 in humans).
- **SINEs (Short Interspersed Nuclear Elements):** Non-autonomous retrotransposons that do not encode their own enzymes and rely on enzymes provided by other retrotransposons (e.g., LINEs) for their transposition. Alu elements in humans are a well-known example.

- **Significance:** Retrotransposons are major drivers of genome evolution, contributing to genome size variation, gene duplication, gene regulation, and the creation of new genes. Their insertional activity can also cause mutations, leading to genetic diseases.

(c) Infective heredity in Paramecium



- **Infective heredity** in *Paramecium aurelia* refers to a classic example of extranuclear inheritance where a stable, inherited trait is conferred by a symbiotic microorganism (or a viral particle) residing in the cytoplasm of the host cell. The most famous example is the inheritance of the "killer" trait in *Paramecium* mediated by **kappa particles**.
- **Killer Trait and Kappa Particles:**
    - Certain strains of *Paramecium aurelia* are known as "killers" because they release a toxic substance called **paramecin** into the surrounding medium, which is lethal to other "sensitive" strains of *Paramecium* that lack the kappa particles.
    - The ability to produce paramecin is directly linked to the presence of **kappa particles** in the cytoplasm of the killer *Paramecium*. Kappa particles are rod-shaped, DNA-containing bacterial symbionts (*Caedibacter taeniospiralis*).
    - Within the kappa particles, there is a specific DNA plasmid (or prophage) that carries the gene for paramecin toxin.

- **Inheritance Pattern:**
    - The inheritance of the killer trait is **cytoplasmic** and **maternally inherited** (though *Paramecium* has complex sexual reproduction involving conjugation).
    - **Conjugation:**
        - When a killer *Paramecium* (containing kappa particles) conjugates with a sensitive *Paramecium* (lacking kappa particles):
            - **Short Conjugation (no cytoplasmic exchange):** If conjugation is brief, only nuclear material is exchanged. The killer *Paramecium* remains killer, and the sensitive *Paramecium* remains sensitive. The offspring will inherit the parental cytoplasmic state.
            - **Long Conjugation (cytoplasmic exchange):** If conjugation is prolonged, there is cytoplasmic bridge formation, and kappa particles can be transferred from the killer to the sensitive cell. In this case, the sensitive *Paramecium* can become a killer, and its progeny will also be killers.
    - **Nuclear Gene Requirement:** For the kappa particles to persist and express the killer trait, the *Paramecium* host must possess at least one dominant nuclear gene, **K**. If the host is homozygous recessive (kk), it cannot maintain the kappa particles, and even if it initially receives them, it will eventually lose them and become sensitive.
- **Significance:**
    - This system provided early and compelling evidence for the existence of **extranuclear inheritance**, demonstrating that genetic information can be carried and passed on through cytoplasmic elements.

- It highlights the complex interactions between nuclear genes, cytoplasmic factors (like organelles), and symbiotic microorganisms in determining the phenotype of an organism.
- It also showcases the role of infectious agents in influencing the heredity of host traits.

(d) Penetrance and Expressivity

- **Penetrance and Expressivity** are two important concepts in genetics that describe the relationship between a genotype and its corresponding phenotype, especially in cases where the relationship is not straightforward. They explain why individuals with the same genotype might not always show the same phenotype, or might show it to different degrees.
- **Penetrance:**
  - **Definition:** Penetrance refers to the proportion of individuals with a particular genotype who actually express the associated phenotype. In other words, it describes "whether" a gene is expressed.
  - **Types:**
    - **Complete Penetrance:** When 100% of individuals with a particular genotype express the corresponding phenotype. For example, if everyone with the dominant allele for Huntington's disease develops the disease, the gene shows complete penetrance.
    - **Incomplete Penetrance:** When less than 100% of individuals with a particular genotype express the associated phenotype. Some individuals with the genotype may not show the phenotype at all.
  - **Example:** Polydactyly (extra fingers or toes) in humans is inherited as an autosomal dominant trait. However, not every individual who inherits the dominant allele for polydactyly will necessarily have extra digits; some may have the genotype but a normal number of digits. If

80 out of 100 people with the polydactyly genotype actually show polydactyly, the penetrance is 80%.

- **Causes of Incomplete Penetrance:** Can be due to environmental factors, the influence of other genes (modifier genes), or random developmental noise.

- **Expressivity:**
  - **Definition:** Expressivity describes the range of phenotypic variation among individuals who all have the same genotype and express the associated phenotype. It describes "to what extent" a gene is expressed.
  - **Types:**
    - **Constant Expressivity:** When all individuals with a particular genotype express the same phenotype to the same degree.
    - **Variable Expressivity:** When individuals with the same genotype express the phenotype to different degrees, or in different ways.
  - **Example:** Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder that shows variable expressivity. Individuals with the NF1 genotype can exhibit a wide range of symptoms, including:
    - Light brown spots on the skin (café-au-lait spots).
    - Multiple benign tumors on the nervous system (neurofibromas).
    - Skeletal abnormalities.
    - Learning disabilities.
    - One affected individual might have only a few café-au-lait spots, while another from the same family with the same genotype might have numerous neurofibromas and severe skeletal problems.

- **Causes of Variable Expressivity:** Similar to penetrance, variable expressivity can be influenced by environmental factors, genetic background (modifier genes), and epigenetic factors.

- **Relationship:** Penetrance is a population measure (all-or-none expression for a given genotype in a population), while expressivity describes individual variability in phenotype among those who *do* express the trait. A trait can have complete penetrance but variable expressivity, or incomplete penetrance with variable expressivity, or complete penetrance with constant expressivity.