3. PRINCIPLES OF INHERITANCE & VARIATION

Single Page Simple Synopsis

Genetics is the branch of biology that studies inheritance (how traits are passed from parents to offspring) and variation (the differences between individuals within a species).

FOUR CORE CONCEPTS:

1. **Basic Terms:**
   - **Gene:** The basic functional unit of heredity, located on chromosomes.
   - **Allele:** Alternative forms of a single gene
     Ex: T for tall and t for dwarf that control contrasting traits.
   - **Genotype & Phenotype:** The genetic makeup of an individual (genotype) and the observable physical expression of those genes (phenotype).
   - **Homozygous & Heterozygous:** Having identical alleles for a trait
     Ex: TT or tt is homozygous. Having different alleles **Ex:**Tt is heterozygous.
2. **Mendelian Discoveries:**
   - **Incomplete Dominance:** Neither allele is completely dominant, resulting in an intermediate phenotype
   - **Co-dominance:** Both alleles are fully and simultaneously expressed
     Ex: ABO blood typing where both A and B proteins are expressed.
   - **Chromosomal Theory of Inheritance** Proposed by Sutton and Boveri, this theory states that chromosomes are the vehicles of genetic heredity, behaving in the same way as Mendelian "factors" or genes during meiosis and fertilization. This was further expanded by T.H. Morgan
3. **Causes of Genetic Variation:**
   - **Mutation:** Sudden, permanent changes in the DNA sequence.
   - **Genetic Recombination:** The exchange of genetic material between homologous chromosomes during meiosis (crossing over), creating new allele combinations.
   - **Sexual Reproduction:** The random fusion of gametes to create unique offspring.
4. **Genetic Disorders:** Alterations in genes or chromosomes can lead to heritable disorders
   - **Mendelian Disorders:** Caused by mutations in a single gene.
     Ex: Hemophilia, Sickle Cell Anemia, Color Blindness.
   - **Chromosomal Disorders:** Caused by an excess, absence, or abnormal arrangement of chromosomes **Ex:** Down Syndrome, Klinefelter's Syndrome.

FRUIT POINTS

1. **Genetics** is the branch of biology that deals with the inheritance and variation of characters from parents to offspring.
2. **Inheritance** is the fundamental process by which characters are passed on from parent to progeny.
3. **Variation** represents the degree by which progeny differ from their parents in their characteristics.
4. **Polygenic traits** are characters that are generally controlled by three or more genes.
5. **Polygenic inheritance** accounts for the influence of the environment alongside genetic factors.
6. **Pleiotropy** occurs when a single gene can exhibit multiple phenotypic expressions.
7. **Phenylketonuria (PKU)** in humans is a disease caused by a single gene mutation.
8. The chromosomal basis of **sex determination** was first suggested by experiments in insects.
9. In 1891, **Henking** identified a specific nuclear structure during spermatogenesis in some insects.
10. Henking called this structure the **X body**, which was later identified as the X-chromosome.
11. **Autosomes** are the chromosomes that are the same in both males and females.
12. Humans have **23 pairs of chromosomes**; 22 pairs are autosomes and one pair consists of sex chromosomes.
13. All human **ova** carry only the X-chromosome.
14. The **genetic makeup of the sperm** (X or Y) determines the sex of the child during fertilization.
15. **Mutation** is a phenomenon that results in alterations of DNA sequences, leading to changes in genotype and phenotype.
16. **Point mutation** involves a change in a single base pair of DNA, such as in sickle cell anemia.
17. **Frame-shift mutations** are caused by the deletion or insertion of base pairs of DNA.
18. **Mendelian disorders** are genetic disorders determined by alterations or mutations in a single gene.
19. **Colour Blindness** is a sex-linked recessive disorder where an individual fails to discriminate between red and green.
20. **Haemophilia** is a sex-linked recessive disease that affects a protein involved in the blood-clotting cascade.

21. In sickle-cell anaemia, **Glutamic acid is substituted by Valine** in the beta globin chain.
22. **Thalassemia** is an autosome-linked recessive blood disease resulting in a reduced synthesis of globin chains.
23. A human female with Turner's syndrome has **45 chromosomes with XO**. **[NEET-2014]**
24. The term 'linkage' was coined by **T.H.Morgan** **[NEET-2015]**
25. A pleiotropic gene **controls multiple traits in an individual**. **[NEET-2015]**
26. Multiple alleles are present **at the same locus of the chromosome**. **[NEET-2015]**
27. Alleles are **different molecular forms of a gene**. **[NEET-2015]**
28. A **disease** caused by an autosomal primary non-disjunction is **Down's syndrome**.**[NEET-2017]**
29. **ESTs (Expressed Sequence Tags)** refers to **genes expressed as RNA.** **[NEET-2019]**
30. **Variations** caused by **mutation** as proposed by Hugo de Vries are **random and directionless** **[NEET-2019]**
31. **The genetic disorder** in which an individual has an overall masculine development gynaecomastia and is sterlie is **Klinefelter's syndrome.** **[NEET-2019]**
32. Experimental verification of the chromosomal theory of inheritance was done by **Morgan**. **[NEET-2020]**
33. The **XO -type** of sex determination can be found in grasshoppers. **[NEET-2022]**
34. Broad palm with single palm crease is visible in a person suffering from Down's syndrome. **[NEET-2023]**
35. **Pleiotropism** refers to a single gene affecting multiple phenotypic expression. **[NEET-2023]**
34. **Non-mendelian inheritance pattern** is the pattern of inheritance for polygenic trait. **[NEET-2025]**