CUET (UG) Biology Notes: Principles of Inheritance and Variation

1. Mendel's Laws of Inheritance (One Gene)

Gregor Mendel conducted hybridization experiments on garden peas (Pisum sativum) for seven years (1856–1863).

Key Concepts & Crosses

- Phenotype: The physical expression of a trait (e.g., Tall or Dwarf).
- Genotype: The genetic makeup of an organism (e.g., TT, Tt, tt).
- Alleles: Genes which code for a pair of contrasting traits.
- Monohybrid Cross: A cross considering only one character. The F_2 generation phenotypic ratio is strictly 3:1 (Tall:Dwarf) and the genotypic ratio is 1:2:1 (TT:Tt:tt).

The Laws

1. Law of Dominance: Characters are controlled by discrete units called factors (genes). In a dissimilar pair of factors, one dominates the other (dominant vs. recessive).
2. Law of Segregation: Alleles do not show any blending. During gamete formation, the factors/alleles of a pair segregate (separate) from each other such that a gamete receives only one of the two factors.
    - Note: This law is universally applicable with no exceptions.

Test Cross & Back Cross

- Test Cross: Crossing an organism with a dominant phenotype (but unknown genotype) with a homozygous recessive parent. Used to determine the unknown genotype. If the offspring are 100% dominant, the parent was homozygous. If they are 50% dominant and 50% recessive (1:1 ratio), the parent was heterozygous.
- Back Cross: Crossing an F_1 hybrid with either of its parents.

2. Deviations from Mendelism

Mendelian ratios are altered when gene interactions occur.

Phenomenon	Description & Examples
Incomplete Dominance	The F_1 phenotype does not resemble either parent but is in-between the two. Phenotypic and genotypic ratios are both 1:2:1. • Example: Flower color in Snapdragon (Antirrhinum majus) or Dog flower. (Red \times White = Pink).
Co-dominance	Both alleles in a heterozygote express themselves completely and simultaneously. • Example: ABO blood grouping in humans.

Multiple Alleles (ABO Blood Groups)

- ABO blood groups are controlled by the gene I.
- The gene has three alleles: I^A, I^B, and i.
- I^A and I^B produce slightly different forms of sugar on the RBC surface and are dominant over i.
- When I^A and I^B are present together, they both express their own types of sugars (Co-dominance), resulting in blood group AB.

Pleiotropy & Polygenic Inheritance

- Pleiotropy: A single gene can exhibit multiple phenotypic expressions. (e.g., Phenylketonuria, where one mutation causes mental retardation, hair reduction, and skin pigmentation changes).
- Polygenic Inheritance: A single trait is controlled by three or more genes. The phenotype reflects the contribution of each allele (quantitative inheritance). (e.g., Human skin color, human height).

3. Inheritance of Two Genes & Chromosomal Theory

Law of Independent Assortment (Dihybrid Cross)

When two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters.

- Dihybrid Phenotypic Ratio: 9:3:3:1 (e.g., Round Yellow : Round Green : Wrinkled Yellow : Wrinkled Green).

Chromosomal Theory of Inheritance

- Proposed independently by Walter Sutton and Theodor Boveri.
- They noted that the behavior of chromosomes is parallel to the behavior of Mendelian genes. Chromosomes, as well as genes, occur in pairs, and the two alleles of a gene pair are located on homologous sites on homologous chromosomes.

Linkage and Recombination

- Discovered by Thomas Hunt Morgan using fruit flies (Drosophila melanogaster).
- Linkage: The physical association of genes on a chromosome. Tightly linked genes show very low recombination.
- Recombination: The generation of non-parental gene combinations (via crossing over).
- CUET Fact: Morgan's student Alfred Sturtevant used the frequency of recombination to map gene positions on chromosomes.

4. Sex Determination Mechanisms

Establishment of sex through genetic/chromosomal means.

System	Mechanism	Organisms
XX-XY Type	Male Heterogamety. Females are XX, Males are XY. Male determines sex of offspring.	Humans, Drosophila
XX-XO Type	Male Heterogamety. Females are XX, Males have only one X (XO).	Grasshoppers, many insects
ZW-ZZ Type	Female Heterogamety. Females are ZW (heterogametic), Males are ZZ (homogametic).	Birds, some reptiles
Haplodiploidy	Sex is determined by the number of sets of chromosomes. Fertilized eggs (2n = 32) become females (queens/workers). Unfertilized eggs develop parthenogenetically into males/drones (n = 16).	Honey bees

5. Genetic Disorders

Pedigree Analysis

A visual tool to trace the inheritance of a specific trait, abnormality, or disease over several generations in a family.

- Squares = Males; Circles = Females.
- Shaded = Affected individual.

Mendelian Disorders (Gene Mutations)

These disorders are mainly determined by alteration or mutation in a single gene.

Disorder	Inheritance Pattern	Key Characteristics
Colour Blindness	Sex-linked (X-linked) Recessive	Inability to distinguish red and green colours. More common in males (requires only one mutant X) than females (requires two).

Haemophilia	Sex-linked (X-linked) Recessive	A single cut results in non-stop bleeding due to a defect in the blood coagulation protein cascade. Queen Victoria was a famous carrier.
Sickle-cell Anaemia	Autosomal Recessive	Substitution of Glutamic acid by Valine at the 6th position of the beta-globin chain. RBCs become sickle-shaped under low oxygen tension. Genotype: Hb^S Hb^S (affected).
Phenylketonuria (PKU)	Autosomal Recessive	Lack of an enzyme that converts the amino acid phenylalanine into tyrosine. Phenylalanine accumulates and converts into phenylpyruvic acid, causing mental retardation.
Thalassemia	Autosomal Recessive	Quantitative problem of synthesizing too few globin molecules, leading to anemia. Alpha-thalassemia is linked to chromosome 16; Beta-thalassemia to chromosome 11.

Chromosomal Disorders

Caused by absence, excess, or abnormal arrangement of one or more chromosomes.

- Aneuploidy: Gain or loss of a chromosome due to failure of segregation of chromatids during cell division (e.g., Down's syndrome).
- Polyploidy: Increase in a whole set of chromosomes due to failure of cytokinesis (common in plants).

Specific Human Chromosomal Syndromes:

1. Down's Syndrome: Trisomy of Chromosome 21 (47 chromosomes). Symptoms include short stature, small round head, furrowed tongue, partially open mouth, and mental retardation.
2. Klinefelter's Syndrome: Presence of an additional copy of X-chromosome in a male (Karyotype: 47, XXY). Individuals are sterile males with overall masculine development but expressed feminine features (e.g., enlarged breasts/gynaecomastia).
3. Turner's Syndrome: Absence of one of the X chromosomes in a female (Karyotype: 45, XO). Individuals are sterile females with rudimentary ovaries and lack of secondary sexual characters.