Genetics Lecture 6

1. Single-Gene Inheritance

Many human traits and diseases are determined primarily by one gene. These traits often follow Mendelian inheritance patterns and can be tracked in families using pedigrees.

2. Importance of Family History

Family history helps predict recurrence risk, identify inheritance patterns, and guide diagnosis of inherited disorders.

3. Degrees of Dominance

Complete dominance: heterozygote has same phenotype as dominant homozygote. Incomplete dominance: heterozygote shows intermediate phenotype. Codominance: both alleles are expressed distinctly.

4. Tay-Sachs Disease

Autosomal recessive disorder caused by deficiency of a lipid-metabolizing enzyme, leading to lipid accumulation in CNS, neurodegeneration, hypotonia, and early death.

5. Dominance Depends on Level

Tay-Sachs can appear recessive clinically, incompletely dominant biochemically (50% enzyme in carriers), and codominant at molecular level because both alleles are expressed.

6. Frequency of Dominant Alleles

Dominant alleles are not always common. Example: polydactyly is dominant but rare compared with normal five digits.

7. Multiple Alleles

A gene may have more than two alleles in the population, though each person carries only two alleles (except sex chromosome exceptions).

8. ABO Blood Group

Three alleles: IA, IB, i. IA and IB are codominant; both dominate i. Genotypes determine A, B, AB, or O blood types.

9. Pleiotropy

One gene affects multiple body systems. Example: cystic fibrosis causes abnormal chloride transport leading to thick mucus, lung disease, pancreatic problems, and infertility.

10. Epistasis

One gene masks or modifies another gene at a different locus. Labrador coat color: B/b determines black or brown pigment, E/e controls pigment deposition. ee causes yellow/white regardless of B gene.

11. Recessive Epistasis Ratio

Classic phenotypic ratio is 9:3:4 in dihybrid crosses.

12. Polygenic Inheritance

Many traits result from additive effects of multiple genes, producing continuous variation. Examples: skin color, height, diabetes risk, hypertension.

13. Multifactorial Traits

Phenotype results from both genes and environment. Example: diabetes risk increases with genetic susceptibility plus obesity and high sugar intake.

14. Norm of Reaction

Range of phenotypes produced by one genotype under different environmental conditions. Example: hydrangea flower color changes with soil pH.

15. Human Genetics Limitations

Humans are difficult research subjects because of long generation time, small family size, and ethical limits on breeding experiments.

16. Pedigree Analysis

Pedigrees use standardized symbols to track traits through generations and estimate inheritance patterns and recurrence risk.

17. Probability Rules

Multiplication rule is used for independent events occurring together; addition rule is used when either of mutually exclusive events may occur.

18. High-Yield

Autosomal recessive diseases often skip generations. Codominance example = ABO. Pleiotropy = one gene, many effects. Epistasis = one gene masks another. Polygenic = many genes, continuous traits.