Genetics and Diseases

1st Course

Lecture : 5

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Definition of Genetics

- Genetics is the branch of biology that studies genes, heredity, and variations in living organisms.

Basic Concepts

1. **Gene:**

- Basic unit of heredity that carries information for a specific trait.

2. **DNA:**

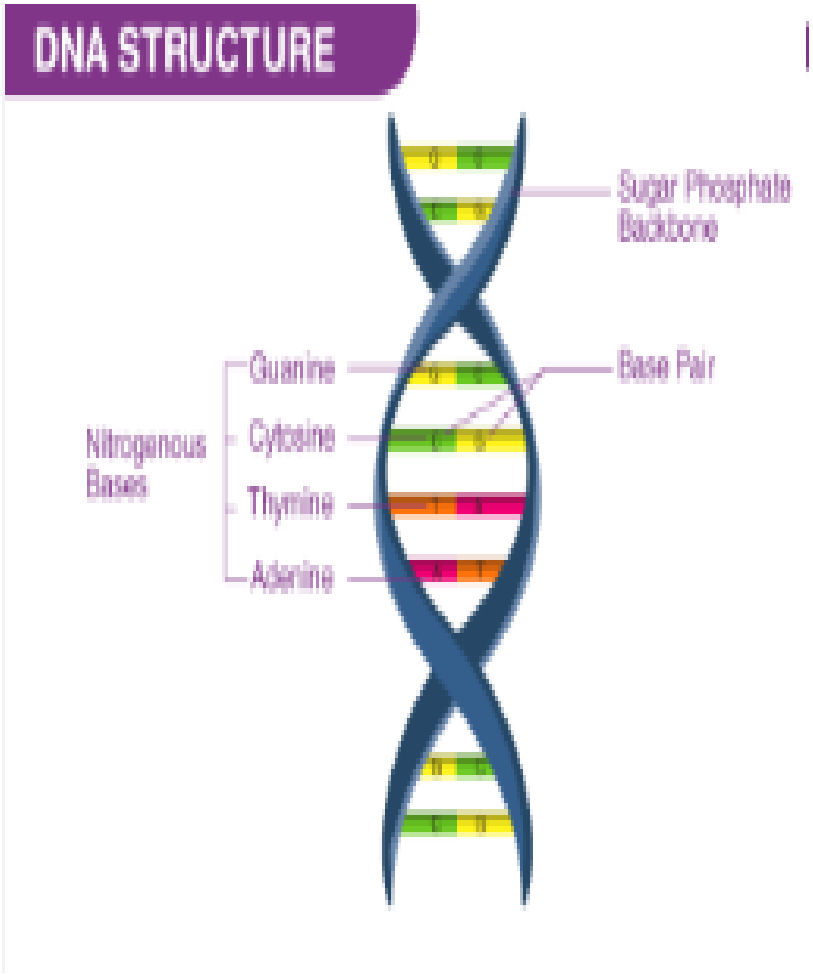
- Deoxyribonucleic acid, the molecule that contains genetic information.

3. **Chromosome:**

- Thread-like structure made of DNA and proteins, located in the cell nucleus.

Structure of DNA

- DNA is a double helix composed of two strands made of nucleotides (adenine, thymine, cytosine, and guanine).
- A pairs with T, and C pairs with G.



Genetic Code and Protein Synthesis

- Genes contain the code for making proteins.
- DNA → mRNA → Protein
- This process is essential for cell function and structure.

Types of Genes

- Dominant Gene:

  Expressed even if only one copy is present.

- Recessive Gene:

  Expressed only when two copies are present.

- Co-dominant Genes:

  Both alleles are expressed equally (e.g., blood groups).

Chromosomes and Karyotype

- Humans have 46 chromosomes (23 pairs).
- Karyotyping is used to study the number and structure of chromosomes.

Mutations

1. A mutation is a change in the DNA sequence.
2. It may cause no effect, or lead to disease.
3. Types: Point mutation, deletion, insertion, duplication.

Genetic Disorders

1. Genetic disorders result from abnormalities in genes or chromosomes.
2. They can be inherited or occur spontaneously.

Single Gene Disorders

- Caused by a mutation in a single gene.
- They follow Mendelian patterns of inheritance (dominant or recessive).

Examples of Single Gene Disorders

1. Cystic Fibrosis: Thick mucus affecting lungs and pancreas.
2. Sickle Cell Anemia: Abnormal hemoglobin causing sickle-shaped red cells.
3. Hemophilia: Deficiency of clotting factors causing bleeding tendency.

Chromosomal Disorders

- Caused by structural or numerical abnormalities of chromosomes.
- Examples:

1. Down Syndrome (Trisomy 21)
2. Turner Syndrome (45, X)
3. Klinefelter Syndrome (47, XXY).

Multifactorial Disorders

- Caused by interaction between genes and environmental factors.
- Examples:

1. Diabetes mellitus
2. Hypertension
3. heart disease.

Genetic Screening and Diagnosis

- Techniques include:

1. Blood tests
2. DNA analysis
3. Chromosomal studies

- Used for early detection and prevention.

Genetic Counseling

- Process of informing individuals and families about genetic risks, inheritance patterns, and testing options.

Prenatal Diagnosis

- Testing during pregnancy to detect genetic disorders.
- Methods:

1. Amniocentesis
2. chorionic villus sampling
3. ultrasound.

Genetic Role in Anesthesia

- Some genetic disorders influence anesthesia response.
- Example:

1. Malignant Hyperthermia
2. Pseudo-cholinesterase deficiency.

Ethical Considerations

- Genetic information must be handled with confidentiality and respect for patient autonomy.

Summary

1. Genetics explains how traits and diseases are inherited.
2. Genetic disorders can result from mutations or chromosomal abnormalities.
3. Understanding genetics helps in diagnosis, prevention, and treatment.