BHARATHIDASAN UNIVERSITY

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Programme: M.Sc., Biomedical Science
(5 Year Integrated Program)

Course Title : Principles of Genetics
Course Code : BM24C4

Unit-II
Patterns of Inheritance and Genetic Analysis

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Patterns of Inheritance and Genetic Analysis in Humans

(Family Studies, Population Genetics, Gene Mapping, and Multifactorial Inheritance)

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Patterns of Inheritance

Refers to the ways genetic traits are transmitted from one generation to the next.

1. **Mendelian Inheritance (Single-Gene Traits):**

   Traits controlled by a single gene following Mendel's laws.

   Example:

   - Dominant: Huntington's disease (caused by a dominant allele).
   - Recessive: Cystic fibrosis (caused by two copies of a recessive allele).

2. **Non-Mendelian Inheritance:**

   Codominance: Both alleles are expressed equally.
   Example: Blood type AB (A and B alleles are codominant).

   Incomplete Dominance: Blending of traits.
   Example: Pink flowers from red and white parents.

   Mitochondrial Inheritance: Traits inherited through mitochondrial DNA.
   Example: Leber's hereditary optic neuropathy.

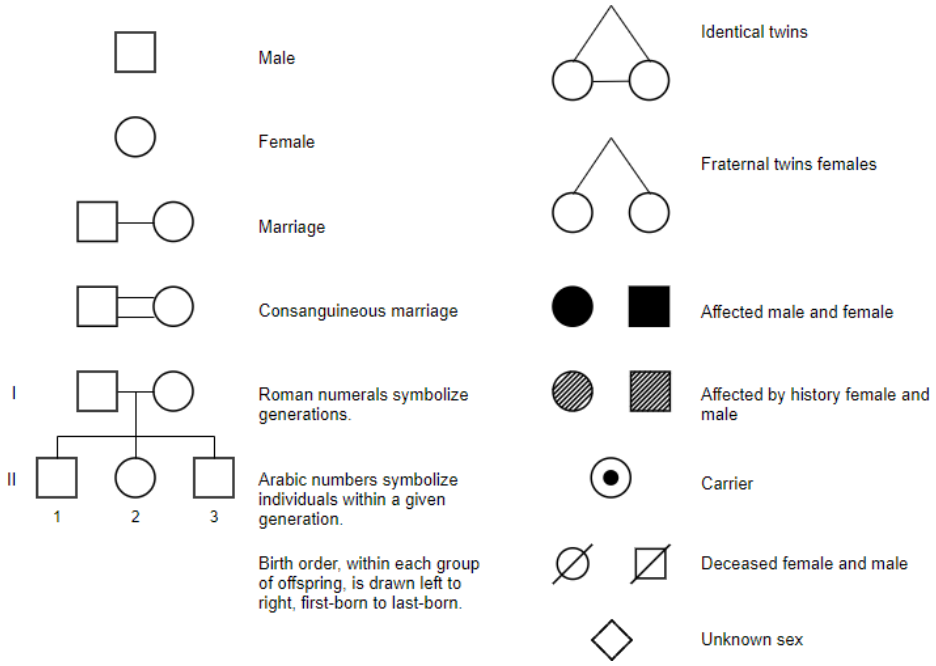
Family Studies in Genetics

Purpose:

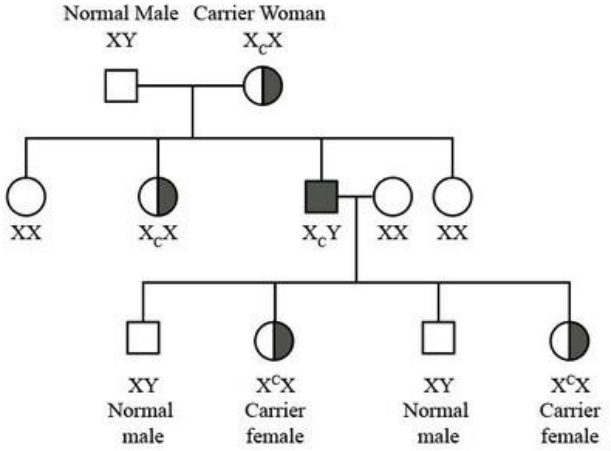
- Trace inheritance patterns of diseases or traits.
- Identify carriers and risks for future generations.

Pedigree Analysis:

- Diagram showing relationships and inheritance of a trait over generations.



Tracking hemophilia in a family tree.



Twin Studies:

- Compare traits in monozygotic (identical) vs dizygotic (fraternal) twins.
  Example: Concordance rates for schizophrenia are higher in monozygotic twins.

Population Genetics

Study of genetic variation within populations and how it changes over time.

1. **Allele Frequencies:** Proportion of a specific allele in a population.

   Example: Sickle-cell allele is more common in malaria-endemic regions.
   Calculated using the Hardy-Weinberg Equation:
   $\mathbf{p^2 + 2pq + q^2 = 1}$, where p and q are allele frequencies.

2. **Gene Polymorphism:**

   Presence of multiple alleles at a locus within a population.
   Example: Polymorphism in the ABO blood group system.

3. **Segregation Analysis:**

   Statistical analysis to determine the mode of inheritance (dominant, recessive, etc.).

4. **Genetic Linkage:**

   Tendency of genes close together on a chromosome to be inherited together.
   Example: Genes for red hair and fair skin are often linked.

Polygenic and Multifactorial Inheritance

Polygenic Traits:

- Traits controlled by multiple genes.
  Example: Height, skin color.

Multifactorial Traits:

- Traits influenced by both genetic and environmental factors.
  Example: Diabetes, heart disease.

Heritability:

- Proportion of trait variation explained by genetic factors (ranges from 0 to 1).
  Example: Heritability of intelligence is estimated at 0.5
  (50% genetic, 50% environmental).

Multifactorial Disorders:

1. Type 2 Diabetes: Genetic predisposition + lifestyle factors (diet, exercise).
2. Heart Disease: Genes affecting cholesterol + risk factors like smoking.
3. Asthma: Interaction of genetic susceptibility and allergens.

Prevention and Management:

Lifestyle changes and personalized medicine based on genetic risk.

Gene Mapping

Process of determining the location of genes on a chromosome

1. **Linkage Mapping :**

   Based on recombination frequencies during meiosis.
   Genes closer together recombine less often.
   Example: Mapping genes for cystic fibrosis to chromosome 7.

2. **Physical Mapping:**

   Measures actual physical distances between genes using molecular techniques (e.g., DNA sequencing).

3. **Applications of Gene Mapping:**

   Identify disease-causing genes (e.g., BRCA1 for breast cancer).
   Develop targeted therapies and gene editing tools (CRISPR).

Identification of Human Disease Genes

Steps in Identifying Disease Genes:

Candidate Gene Approach:

Hypothesize a gene based on known biology.

Example: Huntingtin gene for Huntington's disease.

Genome-Wide Association Studies (GWAS):

Scans entire genome to identify variants associated with diseases.

Example: Identifying risk loci for Alzheimer's disease.

Exome Sequencing:

Focus on protein-coding regions of DNA.

Example: Discovering mutations in rare diseases like Marfan syndrome.

Significance:

Improve diagnosis, predict risk, and develop treatments.

Gene mapping and identification of disease genes are revolutionizing healthcare.