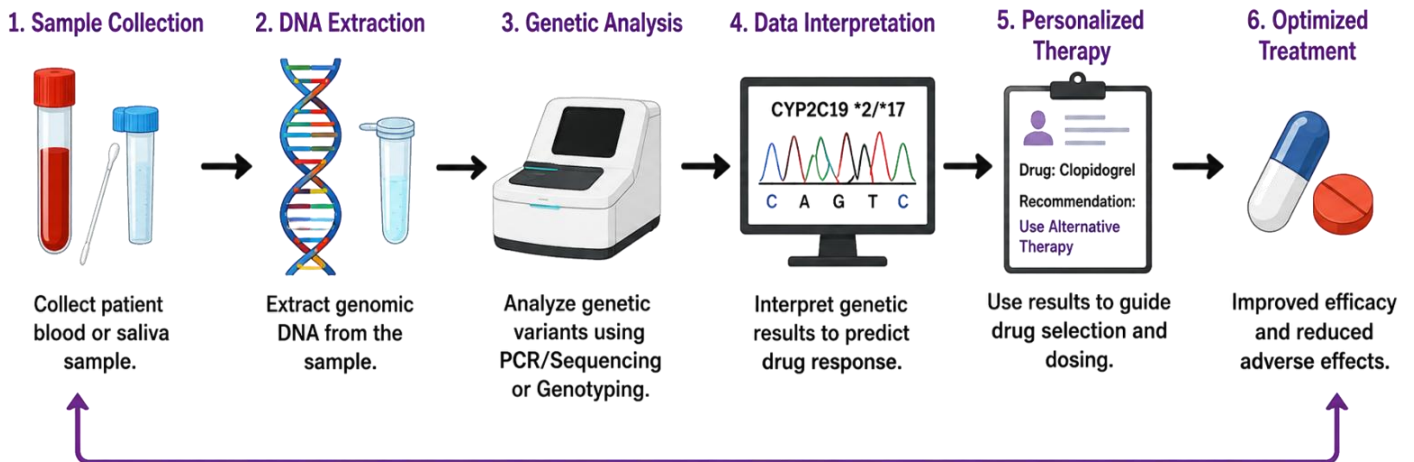


Pharmacogenomics

Introduction to Pharmacogenomics

- Pharmacogenomics is the study of how genes affect an individual's response to drugs.



- It combines pharmacology and genomics to improve drug therapy.
- It helps in selecting the right drug and correct dose for patients.

Basic Principle of Pharmacogenomics

- Genetic differences between individuals influence drug absorption, metabolism, transport, and response.
- Variations in genes can increase or decrease drug effectiveness.

Goals of Pharmacogenomics

- To improve drug safety.
- To reduce adverse drug reactions.
- To increase treatment effectiveness.
- To develop personalized medicine.

Types of Genetic Factors Involved

Types of Genetic Factors Involved



1. Genes Affecting Drug Metabolism

- These genes control enzymes that break down drugs.

2. Genes Affecting Drug Transport

- These genes regulate movement of drugs across cell membranes.

3. Genes Affecting Drug Targets

- These genes influence receptors and proteins that interact with drugs.

Applications of Pharmacogenomics

1. Personalized Medicine

- Drug therapy is tailored according to genetic makeup.

2. Cancer Treatment

- Genetic testing helps choose targeted therapies.

3. Prediction of Drug Toxicity

- High-risk patients can be identified before treatment.

4. Dose Optimization

- Drug doses can be adjusted according to genetic variations.

Advantages of Pharmacogenomics

- It improves treatment outcomes.
- It reduces unwanted side effects.
- It decreases trial-and-error prescribing.
- It supports safer medication use.

Limitations of Pharmacogenomics

1. High cost of genetic testing.
2. Limited availability in some regions.
3. Ethical and privacy concerns.
4. Complex interaction between genes and environment.

Gene Mapping of Disease Gene

- Gene mapping is the process of identifying the location of genes on chromosomes.
- It helps determine genes associated with diseases.

Disease Gene Mapping

- Disease gene mapping identifies genes responsible for inherited disorders.
- It helps understand the genetic basis of diseases.

Types of Gene Mapping

1. Genetic Mapping

- It is based on recombination frequency between genes.

2. Physical Mapping

- It identifies the exact physical location of genes on DNA.

Methods Used in Disease Gene Mapping

1. Linkage Analysis

- It studies inheritance patterns in families.
- Genes located close together tend to be inherited together.

2. Association Studies

- These studies compare genetic variations in affected and healthy individuals.

3. Genome-Wide Association Studies (GWAS)

- The entire genome is analyzed for disease-associated markers.

4. Fluorescence In Situ Hybridization (FISH)

- Fluorescent probes identify gene locations on chromosomes.

Markers Used in Gene Mapping

1. Single Nucleotide Polymorphisms (SNPs)

- These are single base changes in DNA.

2. Microsatellites

- These are short repeated DNA sequences.

Steps in Disease Gene Mapping

1. Collection of family or patient samples.
2. DNA isolation.
3. Analysis of genetic markers.
4. Identification of chromosomal regions linked to disease.
5. Confirmation of disease-causing gene.

Applications of Disease Gene Mapping

- Identification of genetic disorders.
- Prenatal diagnosis.
- Development of targeted therapies.

- Understanding disease mechanisms.

Cloning of Disease Gene

- Cloning of a disease gene is the process of isolating and copying a gene associated with a disease.
- It helps study gene structure and function.

Purpose of Disease Gene Cloning

1. To identify disease-causing mutations.
2. To understand disease mechanisms.
3. To develop diagnostic tests.
4. To support gene therapy research.

Types of Gene Cloning

1. Genomic Cloning

- Entire genomic DNA fragments are cloned.

2. cDNA Cloning

- Complementary DNA prepared from mRNA is cloned.

Steps in Disease Gene Cloning

1. Identification of disease-associated chromosomal region.
2. Isolation of DNA containing the target gene.
3. Cutting DNA using restriction enzymes.
4. Insertion of DNA into a vector.
5. Introduction of recombinant vector into host cells.
6. Selection of cells carrying the cloned gene.
7. Analysis and sequencing of the cloned gene.

Methods Used in Disease Gene Cloning

1. Positional Cloning

- Genes are identified based on chromosomal location.

2. Functional Cloning

- Genes are identified based on protein function.

3. Candidate Gene Approach

- Genes likely involved in disease are specifically studied.

Vectors Used in Gene Cloning

1. Plasmid vectors
2. Bacteriophage vectors
3. BACs and YACs

Applications of Disease Gene Cloning

- Identification of mutations.
- Production of recombinant proteins.
- Development of gene therapy.
- Drug target identification.

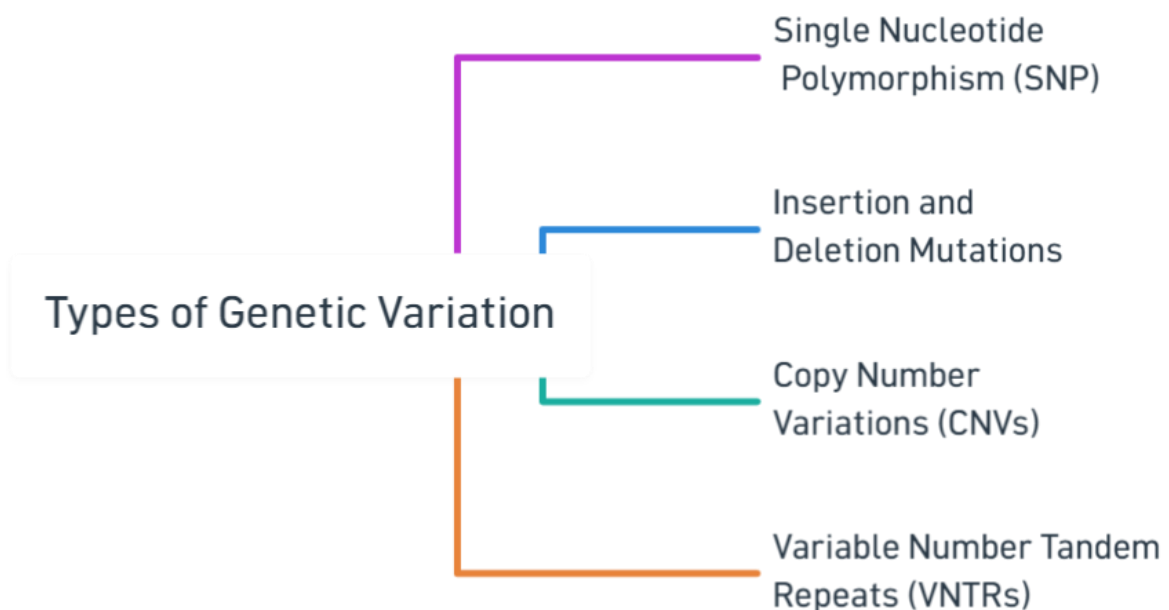
Examples of Cloned Disease Genes

1. CFTR gene in cystic fibrosis.
2. BRCA1 gene in breast cancer.
3. Huntington disease gene.

Genetic Variation and its Role in Health/Pharmacology

- Genetic variation refers to differences in DNA sequences among individuals.
- These variations influence physical traits, disease susceptibility, and drug response.

Types of Genetic Variation



1. Single Nucleotide Polymorphism (SNP)

- A single nucleotide in DNA is changed.
- It is the most common type of genetic variation.



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