FACULTY OF ENGINEERING &TECHNOLOGY
DEPARTMENT OF BIOTECHNOLOGY

Effect of SNP

Silent

Alter the function of the protein

•Directly : alter an amino acid sequence

• indirectly : alter the function of the regulatory sequence

Role of SNPs in Disease predisposition

- **The Common disease are multifactorial**
- **The Genetic differences between human populations make one population more susceptible to particular disease.**

SNPs and Cancer

- **SNPs in genes involved in DNA repair and drug metabolizing enzymes which responsible for metabolism & detoxification of Carcinogens can act as cancer susceptibility genes**

Through

- **Increase activation of chemical carcinogens**

- **Decrease ability of cells to detoxify & repair mutagenic damage**

Methods of identification SNPs

A) Detection of known SNPs

B) Identification of new SNPs

Detection of known SNPs

a) Gel-Based genotyping methods

1 **PCR with restriction enzyme coupled analysis.**

2 **Amplification refractory mutation system (ARMS).**

3-Oligonucleotide ligation assay.

4-Minisequencing.

Detection of known SNPs

b)Non-Gel-based High throughput Genotyping Technologies

1 hybridization using fluorescence resonance energy transfer detection (TaqMan genotyping, Molecular beacons).

2 High-density chip array.

B) Identification of new SNPs

It involves two steps:

1- Conformation-based mutation scanning.

2-Direct DNA sequencing.

Conformation-based mutation scanning

- *Single-strand conformation polymorphism (SSCP).*
- most widely used methods.

Principle:

Single strand DNA tend to fold into complex structure which determines the mobility of the DNA strand in non denaturating gel.

Use and importance of SNPs

- Variations in the DNA sequences of humans can affect how humans develop diseases and respond to pathogens, chemicals, drugs, vaccines, and other agents.
- SNPs are also thought to be key enablers in realizing the concept of personalized medicine

SNP Applications

- Gene discovery and mapping
- Association-based candidate polymorphism testing
- Diagnostics/risk profiling
- Response prediction
- Homogeneity testing/study design
- Gene function identification

Summary

- A **single-nucleotide polymorphism** is a DNA sequence variation occurring when a single nucleotide genome differs between members of a species.

- They can act as biological markers, helping scientists locate genes that are associated with disease. When SNPs occur within a gene or in a regulatory region near a gene, they may play a more direct role in disease by

affecting the gene's function.