



FACULTY OF ENGINEERING & TECHNOLOGY  
DEPARTMENT OF BIOTECHNOLOGY

## Silent

### Alter the function of the protein

- **Directly : alter an amino acid sequence**
- **indirectly : alter the function of the regulatory sequence**



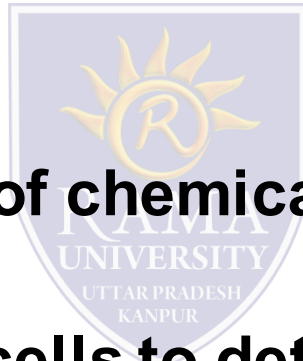
- **The Common disease are multifactorial**
- **The Genetic differences between human populations make one population more susceptible to particular disease.**



- **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

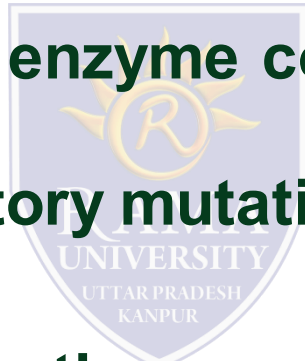
## a) Gel-Based genotyping methods

**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.



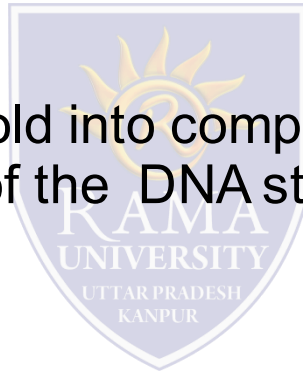


# Conformation-based mutation scanning

- *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 denaturing 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



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



- 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.

