Mutation due to Single Nucleotide Polymorphism

Polymorphism, as related to genomics, refers to the presence of two or more variant forms of a specific DNA sequence that can occur among different individuals or populations. The most common type of polymorphism involves variation at a single nucleotide (also called a single-nucleotide polymorphism, or SNP).

A single nucleotide polymorphism is a genomic variant at a single base position in the DNA. More precisely a variation in a DNA sequence where a single nucleotide (A, C, G, or T) at a specific position in the genome differs between individuals, occurring in at least 1% of a population.

These mutations can occur due to substitution, deletion, or insertion of a single nucleotide

- **Substitution**: When one nucleotide is replaced with another. For example, in the sequence ATAGC, substituting G with C produces ATACC.
- **Deletion**: When a single nucleotide is removed.
- **Insertion**: When a single nucleotide is added.

SNPs can affect gene expression or protein function. They have been linked to multiple human diseases, including viral infections, autoimmune diseases, heart disease, and neurological disorders.

Autoimmune diseases

- **Alopecia areata**: An autoimmune disease that causes bald spots on the scalp and body
- **Crohn's disease**: An autoimmune disease that can be linked to SNPs in the ATG2A gene
- **Rheumatoid arthritis**: An autoimmune disease that can be linked to SNPs
- **Systemic lupus erythematosus (SLE)**: An autoimmune disease that can be linked to SNPs

Heart disease

- SNPs can help predict an individual's risk of developing heart disease
- SNPs can help identify and map complex diseases like heart disease

Neurological disorders

- SNPs in the WIPI4 gene have been linked to neurodegeneration with brain iron accumulation (NBIA) and Rett syndrome
- SNPs in the WIPI2 and WIPI3 genes have been linked to osteoporosis and a neurodevelopmental syndrome
- SNPs in the ATG2A gene have been linked to hyperuricemia and granuloma formation in Crohn's disease

Other diseases linked to SNPs include:

diabetes, schizophrenia, blood pressure, migraine, Alzheimer's disease, and cancer.
SNPs can also help predict how a person will respond to certain drugs.

Factors affecting mutation by SNP

- DNA replication errors
- Environmental exposures (e.g., radiation, chemicals)
- Natural mutation rate
- Specific nucleotide sequence in the region
- Function of the gene where the SNP occurs
- Population genetics factors (e.g., genetic drift, natural selection)
- Activity of DNA repair mechanisms

Findings about the use of SNP in Forensic Purpose from the article

1. Small arrays (approximately 50 loci) of single nucleotide polymorphisms (SNPs) are comparable to short tandem repeat (STR) multiplexes in forensic analysis.
2. A quantitative test is essential for interpreting DNA mixtures
3. SNP arrays are effective in distinguishing between closely related individuals, such as brothers
4. relatively small SNP arrays (50-150 loci) provide likelihood ratios equivalent to STR-based analyses
5. SNP arrays can effectively be used for paternity testing and distinguishing related individuals, except for identical twins.
6. The primary hurdle for forensic SNP applications is developing a balanced multiplex of approximately 50 loci from a low amount of genomic DNA.