



# Methods involved in single nucleotide polymorphism

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## DESCRIPTION

Single Nucleotide Polymorphisms, or SNPs (pronounced "snips"), are the most common type of genetic variation found in humans. Each SNP represents a variation in a single DNA building block known as a nucleotide. SNPs are found naturally throughout a person's DNA. On average, they occur nearly once every 1,000 nucleotides, implying that a person's genome contains 4 to 5 million SNPs (Deng et al., 2011). These variations occur in a large number of people; to be classified as an SNP, a variant must be found in at least 1% of the population. Scientists have discovered over 600 million SNPs in populations all over the world. SNPs are most commonly found in the DNA between genes. They can serve as biological markers, assisting scientists in identifying genes linked to disease. SNPs that occur within a gene or in a regulatory region near a gene may have a more direct impact on disease by affecting gene function (Jia et al., 2013).

The majority of SNPs have no impact on health or development. However, some of these genetic differences have proven to be extremely important in the study of human health. SNPs can predict a person's reaction to certain drugs, susceptibility to environmental factors like toxins, and risk of developing diseases (Jiang et al., 2004). A single nucleotide polymorphism is a difference between individuals in a single position in a DNA sequence. Remember that the DNA sequence is made up of four nucleotide bases: A, C, G, and T. A SNP is defined as a variation in which more than 1% of a population does not carry the same nucleotide at a specific position in the DNA sequence. If an SNP occurs within a gene, the gene is said to have multiple alleles. SNPs may cause variations in the amino acid sequence in these cases. SNPs, on the other hand, are not only associated with genes; they can also be found in non-coding regions of DNA. Although a specific SNP may not cause a disorder, some SNPs are linked to specific diseases.

These associations enable scientists to search for SNPs in order to assess an individual's genetic proclivity to develop a disease (Melson et al., 2014). Furthermore, if certain SNPs are known to be linked to a trait, scientists may examine DNA stretches near these SNPs in an attempt to identify the gene or genes responsible for the trait.

A polynucleotide sequence can have single nucleotides changed (substitution), removed (deletions), or added (insertion). Single nucleotide polymorphisms can occur in gene coding sequences, gene non-coding regions, or gene intergenic regions. Due to the genetic code's degeneracy, SNPs within a coding sequence do not always change the amino acid sequence of the protein that is produced (Xu et al., 2010).

A synonymous SNP (also known as a silent mutation) is one in which both forms produce the same polypeptide sequence; if they produce a different polypeptide sequence, they are nonsynonymous. A nonsynonymous change can be either missense or nonsense, with the former resulting in a different amino acid and the latter in a premature stop codon. SNPs outside of protein-coding regions may still affect gene splicing, transcription factor binding, or the sequence of non-coding Ribonucleic Acid (RNA).

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