



Single-Nucleotide Polymorphism: Genetic Differences Influencing Human Biology and Medication Response

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DESCRIPTION

Single-nucleotide polymorphisms, commonly called SNPs, are variations involving a single nucleotide position within Deoxyribonucleic Acid (DNA). The four basic DNA nucleotides are adenine, thymine, cytosine and guanine. When individuals differ at one nucleotide position, the difference may represent an SNP when it occurs at a sufficient frequency within a population. SNPs are among the most common forms of genetic variation and contribute to differences in physical characteristics, disease susceptibility, biological processes and responses to medicines. A typical SNP involves the replacement of one nucleotide with another. For example, one individual may carry an adenine at a particular DNA position while another may carry guanine. A person can inherit two copies of the same allele or two different alleles at that position. The biological effect depends on the location of the SNP, the gene involved and whether the alteration affects protein production, protein structure, or gene regulation.

SNPs can occur within coding regions of genes. When a nucleotide change alters a codon, it may result in a different amino acid being incorporated into a protein. Such variants are called missense changes. Some substitutions have limited effects, while others can modify protein stability, structure, or activity. A nucleotide alteration can also create a stop signal, resulting in a shortened protein. In contrast, some nucleotide substitutions do not change the amino acid sequence because multiple codons can specify the same amino acid. Many SNPs occur outside protein-coding regions. These locations can include promoters, enhancers, introns and untranslated regions. Although these regions do not directly encode amino acid sequences, they can influence gene expression and Ribonucleic Acid (RNA) processing. An SNP within a regulatory region may increase or decrease the amount of RNA produced from a gene. Changes in RNA levels can subsequently influence the quantity of the corresponding protein within cells.

SNPs can contribute to differences in disease susceptibility. Certain variants are associated with an increased or decreased likelihood of particular disorders. For example, genetic differences can influence lipid metabolism, immune activity, blood pressure regulation and glucose processing. However, the presence of an individual SNP rarely determines disease outcome by itself. Environmental exposure, lifestyle, age, other genetic variants and existing medical conditions can also influence biological outcomes. Pharmacogenomics is an important area in which SNPs have practical relevance. Genetic differences can affect drug-metabolizing enzymes, transport proteins, receptors and other molecules involved in medication action. Variants in *Cytochrome P450 2D6*, *CYP2C9* and other pharmacologically relevant genes can alter the processing of selected medicines. Depending on the medicine and the particular variant, drug exposure may increase or decrease, potentially influencing effectiveness or adverse effects.

CYP2C19 illustrates how an SNP can influence medication response. Some variants reduce enzyme activity, which can change the metabolism of medicines processed through this pathway. Certain antiplatelet medicines require metabolic activation, so reduced enzyme activity may decrease the formation of the active compound. When validated clinical recommendations are available, genotype information can assist healthcare professionals in considering alternative medicines or treatment strategies. SNPs can also affect drug transport. Transporter proteins regulate the movement of medicines through cellular membranes and influence absorption, distribution and elimination. A nucleotide variation within a transporter gene may change protein expression or function. This can modify the concentration of a medicine in blood or tissues and may influence its pharmacological effects.

Immune-related SNPs can influence susceptibility to selected medication reactions. Genetic differences within immune pathways may alter recognition of pharmaceutical compounds or their metabolites. Some genetic markers have been associated

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with serious adverse reactions to specific medicines. Genetic testing can therefore be useful before selected treatments when clinical guidelines recommend a particular test. Laboratories can identify SNPs using several molecular methods. Polymerase chain reaction-based assays can examine specific nucleotide positions, while microarray platforms can assess large collections of known variants. DNA sequencing can provide information across selected genes or much larger genomic regions. The appropriate method depends on the purpose of testing, the number of variants being assessed and the clinical setting.

Bioinformatics plays an important role in interpreting SNP information. Databases can connect variants with gene locations, predicted functional effects, population frequencies

and available clinical evidence. Computational tools can compare genetic profiles and identify relationships between variants and biological characteristics. Proper interpretation is necessary because not every detected SNP has a known clinical effect. Population genetics also uses SNPs to examine genetic diversity. Differences in allele frequencies between populations can provide information about ancestry, migration patterns and adaptation. Population-level variation is important when evaluating genetic associations because a variant that is common in one population may be less frequent in another. Clinical interpretation should therefore consider the population in which genetic evidence was obtained.