

Allele: One of the variant forms of the DNA sequence at a particular locus, or location, on a chromosome. Different alleles can produce variation on inherited characteristics such as hair or eye color. One form of the allele (the dominant one) may be expressed more than the other form (the recessive one). Some alleles may have no direct affect (silent) but may tag genes or other nearby alleles that are causative or contribute to a genetic characteristic, such as a disease or disease susceptibility.

Chromosome: A compact, threadlike organization in the nucleus of human cells that carries the DNA. Each human has two sets of 23 chromosomes, one of each parent.

Gene: A gene is a segment of the DNA molecule that contains the “instructions” of how, when and where our bodies function. These instructions are written by our letter A, T, C, and G. These letter correspond to nucleotides molecules and their combination make up our genetic code.

Genetic Analysis: The study and analysis of genetic variations in a concrete organism.

Genome: The collection of all the genes in the cell of a particular species. In humans, the genome comprises of about 30000 genes.

Enzyme: A protein produced by a living organism, capable of catalyzing a chemical reaction. Almost all processes in a living organism require some form of enzyme to cause the reactions to happen at a sufficient rate to support life.

Risk factor: A factor in an individual’s genetic, physiological, environmental, or socioeconomic state that affects his/her probability of experiencing a particular disease or outcome. For example people with high body mass index are at increased risk of developing diabetes mellitus type II.

Rs: It is the identification number that is used by researchers worldwide to refer to and name the SNPs.

SNP (Single Nucleotide Polymorphism): A SNP is a genetic variation with a letter change in a single nucleotide. SNP provide a type of DNA marker. They occur very frequently in DNA and can be identified very easily by automated methods.