Finding the effect of missense-SNPs on protein structures and functions of HPRT1 using different tools

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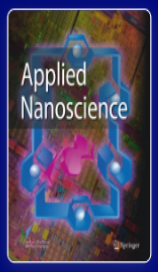

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Abstract

The hypoxanthine–guanine/phosphoribosyl transferase (HGPRT) enzyme is pivotal for purine-nucleotide (IMP) because it catalyzes the transmutation of 6-oxopurinebases to their constituent nucleotides [hypoxanthine to inosine-monophosphate]. In humans, the HPRT1 gene codes for the HGPRT enzyme. LeschNyhan syndrome is an inherited condition induced by a lack of the enzyme-hypoxanthine phosphoribosyl transferase (HPRT). Furthermore, distinguishing harmful nsSNPs with possible disease susceptibility from tolerated mutations is difficult. Various tools and databases are used to anticipate the outcome of mutant genes, then forecast protein stabilization change by one-point mutation, as well as solvent accessibility, secondary-structure, and surface of protein sequence. The 3D structure of a protein is critical for understanding the molecular pathways that cause diseases. The valuation of 3D/structure of the mutant-protein was done by using HOPE in this report. We record the symmetry of 12 possibly deleterious ns-SNPs (L41, S110L, V130D, S104R, G70E, G71R, H204D, D177Y, M143K, G140D, L65F and R48H) in the coding-region of HPRT1 gene.