

Missense mutation or substitution refers to a change in one amino acid in a protein, arising from a point mutation in a single nucleotide. Missense mutation is a type of nonsynonymous substitution in a DNA sequence. Another type of nonsynonymous substitution is a nonsense mutation in which a codon is changed to a premature stop codon that results in truncation of the resulting protein. Missense mutations can render the resulting protein nonfunctional, and such mutations are responsible for human diseases such as Epidermolysis bullosa, sickle-cell disease, and SOD1 mediated ALS.

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Last update: **2024/06/07 02:51**

