RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · SYNONYMOUS

H210= — WFS1 Molecular Atlas Card

Variant type: Synonymous (silent)

Codon: position 210 (Histidine, H) — amino acid unchanged

Domain context: N-terminal cytoplasmic (intrinsically disordered)

SCHEMA CATEGORY: SILENT — SILENT — BUT NEAR AN EXON BOUNDARY (SPLICE EFFECT POSSIBLE)

No amino-acid change (H210 is unchanged), so there is no protein-level structural or stability effect. However, this codon sits within 3 residues of the exon junction near protein position 210 — close enough that the nucleotide change could perturb splicing. Worth a SpliceAI check (Wave 2); otherwise expected to be benign at the protein level.

CLINICAL EVIDENCE

- **Classification:** Conflicting classifications of pathogenicity
- **Review status:** criteria provided, conflicting classifications
- **cDNA change:** c.630C>T
- **ClinVar accession:** VCV001120077
- **Last evaluated:** 2024/06/26 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (synonymous pipeline) on 2026-06-08T02:51:30.599458Z.

WFS1: UniProt O76024, AlphaFold v6. Synonymous variants carry no protein-structural effect.