

P279= — WFS1 Molecular Atlas Card

Variant type: Synonymous (silent)

Codon: position 279 (Proline, P) — 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 (P279 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 280 — 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:** Uncertain significance
- **Review status:** criteria provided, single submitter
- **Associated conditions:** Autosomal dominant nonsyndromic hearing loss 6; WFS1-Related Spectrum Disorders
- **cDNA change:** c.837T>C
- **ClinVar accession:** VCV000907360
- **Last evaluated:** 2018/01/13 00:00
- **Submissions:** 1

Card generated by *wolfram-atlas-batch* (synonymous pipeline) on 2026-06-08T02:51:57.834212Z.

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