

F414= — WFS1 Molecular Atlas Card

Variation type: Synonymous (silent)

Codon: position 414 (Phenylalanine, F) — amino acid unchanged

Domain context: Transmembrane helix 4

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

No amino-acid change (F414 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 412 — 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:** Benign/Likely benign
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Wolfram syndrome 1
- **cDNA change:** c.1242C>T
- **ClinVar accession:** VCV000513249
- **Last evaluated:** 2025/12/15 00:00
- **Submissions:** 1

Card generated by *wolfram-atlas-batch* (synonymous pipeline) on 2026-06-08T02:53:10.532857Z.

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