RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

L377V — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Leucine (L) → Valine (V) at position 377

Domain context: Transmembrane helix 3

ALPHAMISSENSE

- **Pathogenicity score:** 0.3409
- **Class:** ambiguous

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 377:** 84.0

> **DynaMut2 ΔΔG:** not yet computed for this variant — AlphaMissense + AlphaFold

> confidence shown above. Stability ΔΔG and the wild-type/mutant structural

> comparison backfill behind this note.

CLINICAL EVIDENCE

- **Classification:** Uncertain significance
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Inborn genetic diseases
- **cDNA change:** c.1129C>G
- **ClinVar accession:** VCV002195963
- **Last evaluated:** 2025/10/15 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.493025Z.

AlphaMissense (Cheng et al. 2023) · AlphaFold model v6 · UniProt O76024.