RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

L848V — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Leucine (L) → Valine (V) at position 848

Domain context: C-terminal ER-lumenal (calcium binding, calmodulin, chaperone)

ALPHAMISSENSE

- **Pathogenicity score:** 0.1079
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 848:** 83.5

> **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, single submitter
- **cDNA change:** c.2542C>G
- **ClinVar accession:** VCV001317387
- **Last evaluated:** 2020/10/22 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.812724Z.

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