

V348L — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Valine (V) → Leucine (L) at position 348

Domain context: Transmembrane helix 2

ALPHAMISSENSE

- **Pathogenicity score:** 0.1411
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 348:** 89.12

> **DynaMut2 $\Delta\Delta G$:** not yet computed for this variant — AlphaMissense + AlphaFold

> confidence shown above. Stability $\Delta\Delta G$ and the wild-type/mutant structural

> comparison backfill behind this note.

CLINICAL EVIDENCE

- **Classification:** Uncertain significance
 - **Review status:** no assertion criteria provided
 - **Associated conditions:** Optic atrophy
 - **cDNA change:** c.1042G>C
 - **ClinVar accession:** VCV003250165
 - **Last evaluated:** 2023/01/01 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.474987Z.

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