

# L548V — WFS1 Molecular Atlas Card

**Variant type:** Missense

**Substitution:** Leucine (L) → Valine (V) at position 548

**Domain context:** Transmembrane helix 8

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.2068
- **Class:** likely benign

## ALPHAFOLD CONFIDENCE

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- **pLDDT at residue 548:** 85.94

> **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.

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## CLINICAL EVIDENCE

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- **Classification:** Uncertain significance
  - **Review status:** criteria provided, single submitter
  - **Associated conditions:** Autosomal dominant nonsyndromic hearing loss 6; Cataract 41; Wolfram syndrome 1; Wolfram-like syndrome; Type 2 diabetes mellitus
  - **cDNA change:** c.1642C>G
  - **ClinVar accession:** VCV003590704
  - **Last evaluated:** 2023/12/27 00:00
  - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.612251Z.

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