

L514V — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Leucine (L) → Valine (V) at position 514

Domain context: Transmembrane helix 7

ALPHAMISSENSE

- **Pathogenicity score:** 0.1756
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 514:** 88.12

> **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:** 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.1540C>G
 - **ClinVar accession:** VCV003590699
 - **Last evaluated:** 2024/05/30 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.590266Z.

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