RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

V333I — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Valine (V) → Isoleucine (I) at position 333

Domain context: Cytoplasmic loop 1

ALPHAMISSENSE

- **Pathogenicity score:** 0.0465
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 333:** 71.5

> **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:** Benign/Likely benign
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** WFS1-Related Spectrum Disorders; Wolfram syndrome 1; Wolfram-like syndrome; Type 2 diabetes mellitus; Autosomal dominant nonsyndromic hearing loss 6; Cataract 41
- **cDNA change:** c.997G>A
- **ClinVar accession:** VCV000045463
- **Last evaluated:** 2026/02/04 00:00
- **Submissions:** 2

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.465645Z.

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