

I319F — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Isoleucine (I) → Phenylalanine (F) at position 319

Domain context: Transmembrane helix 1

ALPHAMISSENSE

- **Pathogenicity score:** 0.2518
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 319:** 60.62

> **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/Uncertain risk allele
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Wolfram syndrome 1; Cataract 41; Wolfram-like syndrome; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Inborn genetic diseases
- **cDNA change:** c.955A>T
- **ClinVar accession:** VCV000872209
- **Last evaluated:** 2025/05/25 00:00
- **Submissions:** 1

Card generated by wolfram-atlas-batch (missense AlphaMissense mint) on 2026-06-08T02:27:33.454366Z.

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