RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

I489L — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Isoleucine (I) → Leucine (L) at position 489

Domain context: Lumenal loop 3

ALPHAMISSENSE

- **Pathogenicity score:** 0.0569
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 489:** 79.12

> **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:** Likely benign
- **Review status:** criteria provided, single submitter
- **Associated conditions:** Inborn genetic diseases
- **cDNA change:** c.1465A>C
- **ClinVar accession:** VCV004201390
- **Last evaluated:** 2025/08/25 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.571567Z.

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