

V493A — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Valine (V) → Alanine (A) at position 493

Domain context: Lumenal loop 3

ALPHAMISSENSE

- **Pathogenicity score:** 0.137
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 493:** 80.88

> **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, multiple submitters, no conflicts
- **Associated conditions:** Inborn genetic diseases; Wolfram-like syndrome; Cataract 41; Wolfram syndrome 1; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus
- **cDNA change:** c.1478T>C
- **ClinVar accession:** VCV001331576
- **Last evaluated:** 2026/01/26 00:00
- **Submissions:** 1

Card generated by wolfram-atlas-batch (missense AlphaMissense mint) on 2026-06-08T02:27:33.574476Z.

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