RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

E776V — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Glutamic acid (E) → Valine (V) at position 776

Domain context: C-terminal ER-lumenal (calcium binding, calmodulin, chaperone)

ALPHAMISSENSE

- **Pathogenicity score:** 0.9702
- **Class:** likely pathogenic

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 776:** 93.19

> **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:** Monogenic diabetes; WFS1-Related Spectrum Disorders; Nonsyndromic genetic hearing loss; Autosomal dominant nonsyndromic hearing loss 6
- **cDNA change:** c.2327A>T
- **ClinVar accession:** VCV000166606
- **Last evaluated:** 2026/02/01 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.760179Z.

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