RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

A677T — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Alanine (A) → Threonine (T) at position 677

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

ALPHAMISSENSE

- **Pathogenicity score:** 0.4876
- **Class:** ambiguous

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 677:** 81.19

> **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:** Uncertain significance/Uncertain risk allele
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Retinal dystrophy; Childhood onset hearing loss; Cataract 41; Wolfram syndrome 1; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Wolfram-like syndrome
- **cDNA change:** c.2029G>A
- **ClinVar accession:** VCV000198834
- **Last evaluated:** 2025/09/08 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.700824Z.

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