RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

R676C — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Arginine (R) → Cysteine (C) at position 676

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

ALPHAMISSENSE

- **Pathogenicity score:** 0.4861
- **Class:** ambiguous

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 676:** 81.12

> **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:** Uncertain significance
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Cataract 41; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Wolfram syndrome 1; Wolfram-like syndrome; WFS1-Related Spectrum Disorders
- **cDNA change:** c.2026C>T
- **ClinVar accession:** VCV000349321
- **Last evaluated:** 2026/01/24 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.698027Z.

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