

R375H — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Arginine (R) → Histidine (H) at position 375

Domain context: Transmembrane helix 3

ALPHAMISSENSE

- **Pathogenicity score:** 0.1162
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 375:** 83.0

> **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:** Monogenic diabetes; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Cataract 41; Wolfram syndrome 1; Wolfram-like syndrome; Retinal dystrophy
- **cDNA change:** c.1124G>A
- **ClinVar accession:** VCV000504709
- **Last evaluated:** 2026/01/15 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.491455Z.

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