

# R383C — WFS1 Molecular Atlas Card

**Variant type:** Missense

**Substitution:** Arginine (R) → Cysteine (C) at position 383

**Domain context:** Transmembrane helix 3

---

## ALPHAMISSENSE

---

- **Pathogenicity score:** 0.1309
- **Class:** likely benign

## ALPHAFOLD CONFIDENCE

---

- **pLDDT at residue 383:** 82.88

> **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:** Autosomal dominant nonsyndromic hearing loss 6; WFS1-Related Spectrum Disorders; Cataract 41; Wolfram syndrome 1; Type 2 diabetes mellitus; Wolfram-like syndrome
- **cDNA change:** c.1147C>T
- **ClinVar accession:** VCV000904089
- **Last evaluated:** 2026/01/21 00:00
- **Submissions:** 1

---

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.496456Z.

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