

# R147Q — WFS1 Molecular Atlas Card

**Variant type:** Missense

**Substitution:** Arginine (R) → Glutamine (Q) at position 147

**Domain context:** N-terminal cytoplasmic (intrinsically disordered)

---

## ALPHAMISSENSE

---

- **Pathogenicity score:** 0.0934
- **Class:** likely benign

## ALPHAFOLD CONFIDENCE

---

- **pLDDT at residue 147:** 92.5

> **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:** Wolfram syndrome 1; Cataract 41; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Wolfram-like syndrome
  - **cDNA change:** c.440G>A
  - **ClinVar accession:** VCV001386944
  - **Last evaluated:** 2025/08/15 00:00
  - **Submissions:** 1
-

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.373609Z.

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