RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

R138H — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Arginine (R) → Histidine (H) at position 138

Domain context: N-terminal cytoplasmic (intrinsically disordered)

ALPHAMISSENSE

- **Pathogenicity score:** 0.1781
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 138:** 89.06

> **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:** Inborn genetic diseases; Wolfram syndrome 1; Wolfram-like syndrome; Cataract 41; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus
- **cDNA change:** c.413G>A
- **ClinVar accession:** VCV002073332
- **Last evaluated:** 2024/06/09 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.370564Z.

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