

# R517H — WFS1 Molecular Atlas Card

**Variation type:** Missense

**Substitution:** Arginine (R) → Histidine (H) at position 517

**Domain context:** Transmembrane helix 7

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.1344
- **Class:** likely benign

## ALPHAFOLD CONFIDENCE

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- **pLDDT at residue 517:** 87.81

> **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.

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## CLINICAL EVIDENCE

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- **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-like syndrome; Wolfram syndrome 1
  - **cDNA change:** c.1550G>A
  - **ClinVar accession:** VCV001403563
  - **Last evaluated:** 2025/11/25 00:00
  - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.591854Z.

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