

# Y513S — WFS1 Molecular Atlas Card

**Variant type:** Missense

**Substitution:** Tyrosine (Y) → Serine (S) at position 513

**Domain context:** Transmembrane helix 7

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.4652
- **Class:** ambiguous

## ALPHAFOLD CONFIDENCE

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- **pLDDT at residue 513:** 89.88

> **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.

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## CLINICAL EVIDENCE

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- **Classification:** Uncertain significance
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** WFS1-Related Spectrum Disorders; Cataract 41; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Wolfram syndrome 1; Wolfram-like syndrome
- **cDNA change:** c.1538A>C
- **ClinVar accession:** VCV000215412
- **Last evaluated:** 2025/08/27 00:00
- **Submissions:** 1

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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.589450Z.

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