RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

L201M — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Leucine (L) → Methionine (M) at position 201

Domain context: N-terminal cytoplasmic (intrinsically disordered)

ALPHAMISSENSE

- **Pathogenicity score:** 0.3146
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 201:** 73.38

> **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/Uncertain risk allele
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Inborn genetic diseases; Wolfram syndrome 1
- **cDNA change:** c.601C>A
- **ClinVar accession:** VCV000618495
- **Last evaluated:** 2025/06/12 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.396065Z.

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