

A243T — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Alanine (A) → Threonine (T) at position 243

Domain context: N-terminal cytoplasmic (intrinsically disordered)

ALPHAMISSENSE

- **Pathogenicity score:** 0.1094
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 243:** 86.19

> **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/Uncertain risk allele
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Cataract 41; Wolfram-like syndrome; Wolfram syndrome 1; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Inborn genetic diseases
- **cDNA change:** c.727G>A
- **ClinVar accession:** VCV000809616
- **Last evaluated:** 2025/06/10 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.417012Z.

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