

E751K — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Glutamic acid (E) → Lysine (K) at position 751

Domain context: C-terminal ER-lumenal (calcium binding, calmodulin, chaperone)

ALPHAMISSENSE

- **Pathogenicity score:** 0.0955
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 751:** 78.06

> **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
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Inborn genetic diseases; Cataract 41; Autosomal dominant nonsyndromic hearing loss 6; Wolfram syndrome 1; Type 2 diabetes mellitus; Wolfram-like syndrome
- **cDNA change:** c.2251G>A
- **ClinVar accession:** VCV001198372
- **Last evaluated:** 2025/03/17 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.742055Z.

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