

E753K — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Glutamic acid (E) → Lysine (K) at position 753

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

ALPHAMISSENSE

- **Pathogenicity score:** 0.4672
- **Class:** ambiguous

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 753:** 83.19

> **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
 - **Review status:** criteria provided, single submitter
 - **cDNA change:** c.2257G>A
 - **ClinVar accession:** VCV003363735
 - **Last evaluated:** 2023/11/02 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.744309Z.

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