

# A750V — WFS1 Molecular Atlas Card

**Variant type:** Missense

**Substitution:** Alanine (A) → Valine (V) at position 750

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

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.0915
- **Class:** likely benign

## ALPHAFOLD CONFIDENCE

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- **pLDDT at residue 750:** 73.12

> **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, single submitter
  - **cDNA change:** c.2249C>T
  - **ClinVar accession:** VCV002842594
  - **Last evaluated:** 2023/03/16 00:00
  - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.741677Z.

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