

E550V — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Glutamic acid (E) → Valine (V) at position 550

Domain context: Transmembrane helix 8

ALPHAMISSENSE

- **Pathogenicity score:** 0.4497
- **Class:** ambiguous

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 550:** 85.0

> **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.1649A>T
 - **ClinVar accession:** VCV001315759
 - **Last evaluated:** 2019/10/04 00:00
 - **Submissions:** 1
-

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.614183Z.

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