

N849D — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Asparagine (N) → Aspartic acid (D) at position 849

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

ALPHAMISSENSE

- **Pathogenicity score:** 0.2127
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 849:** 83.0

> **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.2545A>G
 - **ClinVar accession:** VCV002908409
 - **Last evaluated:** 2023/08/11 00:00
 - **Submissions:** 1
-

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.813176Z.

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