

# D713N — WFS1 Molecular Atlas Card

**Variation type:** Missense

**Substitution:** Aspartic acid (D) → Asparagine (N) at position 713

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

---

## ALPHAMISSENSE

---

- **Pathogenicity score:** 0.1878
- **Class:** likely benign

---

## ALPHAFOLD CONFIDENCE

---

- **pLDDT at residue 713:** 85.31

> **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.2137G>A
  - **ClinVar accession:** VCV002176101
  - **Last evaluated:** 2024/04/16 00:00
  - **Submissions:** 1
-

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.720409Z.

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