

# D713H — WFS1 Molecular Atlas Card

**Variation type:** Missense

**Substitution:** Aspartic acid (D) → Histidine (H) at position 713

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

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.4445
- **Class:** ambiguous

## ALPHAFOLD CONFIDENCE

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- **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.

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## CLINICAL EVIDENCE

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- **Classification:** Uncertain significance
  - **Review status:** criteria provided, single submitter
  - **cDNA change:** c.2137G>C
  - **ClinVar accession:** VCV001398188
  - **Last evaluated:** 2024/09/19 00:00
  - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.720041Z.

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