

D713G — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Aspartic acid (D) → Glycine (G) at position 713

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

ALPHAMISSENSE

- **Pathogenicity score:** 0.4097
- **Class:** ambiguous

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, multiple submitters, no conflicts
- **Associated conditions:** Cataract 41; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Wolfram syndrome 1; Wolfram-like syndrome; WFS1-Related Spectrum Disorders
- **cDNA change:** c.2138A>G
- **ClinVar accession:** VCV000349322
- **Last evaluated:** 2024/12/02 00:00
- **Submissions:** 1

Card generated by wolfram-atlas-batch (missense AlphaMissense mint) on 2026-06-08T02:27:33.719662Z.

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