RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · MISSENSE

E795D — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Glutamic acid (E) → Aspartic acid (D) at position 795

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

ALPHAMISSENSE

- **Pathogenicity score:** 0.075
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 795:** 51.81

> **DynaMut2 ΔΔG:** not yet computed for this variant — AlphaMissense + AlphaFold

> confidence shown above. Stability ΔΔG 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:** Type 2 diabetes mellitus; Wolfram syndrome 1; Monogenic diabetes; Inborn genetic diseases; Autosomal dominant nonsyndromic hearing loss 6; Cataract 41; Wolfram-like syndrome
- **cDNA change:** c.2385G>C
- **ClinVar accession:** VCV000166609
- **Last evaluated:** 2025/12/10 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.777528Z.

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