RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · IN-FRAME INDEL

c.2390_2391insACG — WFS1 Molecular Atlas Card

Variant type: In-frame indel

Change: 1 residue(s) inserted in frame at position 797

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

SCHEMA CATEGORY: I2 — SINGLE-RESIDUE DELETION IN A SOLUBLE DOMAIN — VARIABLE IMPACT

A single residue removed in C-terminal ER-lumenal (calcium binding, calmodulin, chaperone) (a soluble, non-membrane region) may be tolerated or may locally distort the domain. Worth pharmacological-chaperone exploration if AlphaFold predicts a near-native fold. Predicted structure pending (ColabFold).

STRUCTURAL PREDICTION

- **Reading frame:** preserved (in-frame) — no premature stop, NMD does not apply.
- **Affected domain:** C-terminal ER-lumenal (calcium binding, calmodulin, chaperone)
- **Predicted modified structure:** _pending — AlphaFold/ColabFold prediction of the

modified sequence and backbone-RMSD vs wild-type backfill here (Wave 2)._

CLINICAL EVIDENCE

- **Classification:** Conflicting classifications of pathogenicity
- **Review status:** criteria provided, conflicting classifications
- **Associated conditions:** Wolfram syndrome 1
- **cDNA change:** c.2390_2391insACG

- **ClinVar accession:** VCV000217514
- **Last evaluated:** 2014/08/05 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` *(in-frame indel pipeline) on 2026-06-08T02:41:46.696689Z.*

Schema: `reference/card` `schemaextension.md` *(I1–I3). WFS1: UniProt O76024, AlphaFold v6.*