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

c.2563_2565del — WFS1 Molecular Atlas Card

Variant type: In-frame indel

Change: 1 residue(s) deleted in frame at position 855

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

Not found in the cached WFS1 ClinVar set (`reference/WFS1clinvarvariants.csv` *).*_

Card generated by `wolfram-atlas-batch` *(in-frame indel pipeline) on 2026-06-08T02:41:56.528299Z.*

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