

c.682_684del — WFS1 Molecular Atlas Card

Variant type: In-frame indel

Change: 1 residue(s) deleted in frame at position 228

Domain context: N-terminal cytoplasmic (intrinsically disordered)

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

A single residue removed in N-terminal cytoplasmic (intrinsically disordered) (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:** N-terminal cytoplasmic (intrinsically disordered)
- **Predicted modified structure:** _pending — AlphaFold/ColabFold prediction of the

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

Not found in the cached WFS1 ClinVar set ([reference/WFS1clinvarvariants.csv](#))._
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Card generated by `wolfram-atlas-batch` (in-frame indel pipeline) on 2026-06-08T02:42:06.254545Z.

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