

c.994_996dup — WFS1 Molecular Atlas Card

Variant type: In-frame indel

Change: 1 residue(s) deleted in frame at position 332

Domain context: Cytoplasmic loop 1

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

A single residue removed in Cytoplasmic loop 1 (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:** Cytoplasmic loop 1
- **Predicted modified structure:** _pending — AlphaFold/ColabFold prediction of the modified sequence and backbone-RMSD vs wild-type backfill here (Wave 2)._
[backfill here \(Wave 2\).](#)

CLINICAL EVIDENCE

- **Classification:** Uncertain significance
 - **Review status:** criteria provided, single submitter
 - **cDNA change:** c.994_996dup
 - **ClinVar accession:** VCV002763322
 - **Last evaluated:** 2024/02/02 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (in-frame indel pipeline) on 2026-06-08T02:42:12.047853Z.

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