

c.1504_1527dup — WFS1 Molecular Atlas Card

Variant type: In-frame indel

Change: 8 residue(s) deleted in frame at position 502

Domain context: Transmembrane helix 7

SCHEMA CATEGORY: I3 — MULTI-RESIDUE IN-FRAME INDEL — LIKELY MAJOR STRUCTURAL DISRUPTION

8 residues removed in frame around position 502 (Transmembrane helix 7). A change this size usually perturbs local packing and can propagate to the fold. Gene therapy is the primary path unless an AlphaFold prediction of the modified sequence shows a surprisingly intact fold. Predicted structure pending (ColabFold).

STRUCTURAL PREDICTION

- **Reading frame:** preserved (in-frame) — no premature stop, NMD does not apply.
- **Affected domain:** Transmembrane helix 7
- **Predicted modified structure:** _pending — AlphaFold/ColabFold prediction of the modified sequence and backbone-RMSD vs wild-type backfill here (Wave 2)._
[View structure](#)

CLINICAL EVIDENCE

- **Classification:** Likely pathogenic
- **Review status:** no assertion criteria provided
- **Associated conditions:** WFS1-related disorder
- **cDNA change:** c.1504_1527dup
- **ClinVar accession:** VCV003345213
- **Last evaluated:** 2024/07/16 00:00

- **Submissions: 1**
-

Card generated by `wolfram-atlas-batch` (in-frame indel pipeline) on 2026-06-08T02:41:19.576136Z.

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