RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · COPY-NUMBER

4p16.3-14 duplication — WFS1 Molecular Atlas Card

Variant type: Copy-number variant (duplication)

Region: Chromosome 4p16.3-14 (gene-level structural event)

CNV-GAIN — COPY-NUMBER DUPLICATION — 4P16.3-14

This is a large copy-number duplication spanning 4p16.3-14 — a gene-level event causing an extra copy of this region (gene over-dosage / partial trisomy). CNVs act by dosage rather than by altering a single residue, so AlphaFold residue mapping, ΔΔG, NMD and AlphaMissense don't apply. Therapeutically these are gene-replacement candidates (restore a working copy); per-residue chaperone/readthrough strategies are not relevant. Confirm breakpoints and gene content against the ClinVar record below.

CLINICAL EVIDENCE

- **Classification:** Pathogenic
- **Review status:** no assertion criteria provided
- **Associated conditions:** See cases
- **ClinVar accession:** VCV000144195
- **Genomic variant:** GRCh38/hg38 4p16.3-14(chr4:72555-39477144)x3
- **Last evaluated:** 2010/05/27 00:00

Card generated by `wolfram-atlas-batch` (CNV pipeline) on 2026-06-08T03:03:52.350716Z.

CNVs are gene-level events; WFS1 protein reference UniProt O76024 is not residue-mapped here.