RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · COPY-NUMBER

4p16.3-16.1 deletion — WFS1 Molecular Atlas Card

Variant type: Copy-number variant (deletion)

Region: Chromosome 4p16.3-16.1 (gene-level structural event)

CNV-LOSS — COPY-NUMBER DELETION — 4P16.3-16.1

This is a large copy-number deletion spanning 4p16.3-16.1 — a gene-level event causing loss of one copy of this region (wolframin haploinsufficiency or, with a second hit, loss of function). 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:** criteria provided, single submitter
- **Associated conditions:** See cases
- **ClinVar accession:** VCV000057936
- **Genomic variant:** GRCh38/hg38 4p16.3-16.1(chr4:85149-7063699)x1
- **Last evaluated:** 2011/08/12 00:00

Card generated by `wolfram-atlas-batch` (CNV pipeline) on 2026-06-08T03:03:52.352841Z.

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