

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, $\Delta\Delta G$, 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:** VCV000252966
 - **Genomic variant:** GRCh37/hg19 4p16.3-16.1(chr4:49450-8872474)x1
 - **Last evaluated:** 2016/05/03 00:00
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Card generated by wolfram-atlas-batch (CNV pipeline) on 2026-06-08T03:03:52.344396Z.

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