

4p16.3-15.33 deletion — WFS1 Molecular Atlas Card

Variant type: Copy-number variant (deletion)

Region: Chromosome 4p16.3-15.33 (gene-level structural event)

CNV-LOSS — COPY-NUMBER DELETION — 4P16.3-15.33

This is a large copy-number deletion spanning 4p16.3-15.33 — 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:** not provided
 - **ClinVar accession:** VCV004796162
 - **Genomic variant:** GRCh38/hg38 4p16.3-15.33(chr4:68454-12774004)x1
 - **Last evaluated:** 2025/12/10 00:00
-

Card generated by wolfram-atlas-batch (CNV pipeline) on 2026-06-08T03:03:52.327191Z.

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