

# 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:** no assertion criteria provided
  - **Associated conditions:** not provided
  - **ClinVar accession:** VCV000685188
  - **Genomic variant:** GRCh37/hg19 4p16.3-15.33(chr4:3374195-13468480)x1
  - **Last evaluated:** 2018/01/17 00:00
- 

*Card generated by wolfram-atlas-batch (CNV pipeline) on 2026-06-08T03:03:52.338915Z.*

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