

# 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)

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

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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.

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## CLINICAL EVIDENCE

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- **Classification:** Pathogenic
  - **Review status:** criteria provided, single submitter
  - **Associated conditions:** See cases
  - **ClinVar accession:** VCV000057938
  - **Genomic variant:** GRCh38/hg38 4p16.3-16.1(chr4:85149-7843616)x1
  - **Last evaluated:** 2011/08/12 00:00
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*Card generated by wolfram-atlas-batch (CNV pipeline) on 2026-06-08T03:03:52.352126Z.*

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