

# 4p16.3-12 duplication — WFS1 Molecular Atlas Card

**Variant type:** Copy-number variant (duplication)

**Region:** Chromosome 4p16.3-12 (gene-level structural event)

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## CNV-GAIN — COPY-NUMBER DUPLICATION — 4P16.3-12

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This is a large copy-number duplication spanning 4p16.3-12 — a gene-level event causing an extra copy of this region (gene over-dosage / partial trisomy). 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:** no assertion criteria provided
  - **Associated conditions:** See cases
  - **ClinVar accession:** VCV000148186
  - **Genomic variant:** GRCh38/hg38 4p16.3-12(chr4:36424-47491595)x3
  - **Last evaluated:** 2011/03/21 00:00
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Card generated by *wolfram-atlas-batch* (CNV pipeline) on 2026-06-08T03:03:52.347950Z.

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