

c.460+16G_T — WFS1 Molecular Atlas Card

Variant type: Splice site

Boundary: donor (5' splice site) · intronic offset +16

Nearest protein position: ~154 (N-terminal cytoplasmic (intrinsically disordered))

SCHEMA CATEGORY: S3 — CRYPTIC DONOR ACTIVATION PREDICTED (SPliceAI ΔS 0.21)

SpliceAI's strongest signal is a NEW cryptic donor site (ΔS 0.21, ~-6 nt from the variant; acceptor-gain 0.00, acceptor-loss 0.00, donor-gain 0.21, donor-loss 0.00). Cryptic activation produces a non-canonical junction whose reading-frame impact depends on the exact cryptic position — typically a partial exon gain/loss. Outcome is context-dependent; wet-lab RNA validation required. Flagged S3.

SPLICE PREDICTION

- **Affected site:** donor (5' splice site), extended splice region
- **SpliceAI delta scores** (GRCh38 chr4:6289147 G>T):
 - acceptor gain **0.00** · acceptor loss **0.00**
 - donor gain **0.21** · donor loss **0.00**
- **Predicted outcome:** Cryptic donor activation predicted (SpliceAI ΔS 0.21)

CLINICAL EVIDENCE

- **Classification:** Likely benign
- **Review status:** criteria provided, single submitter
- **cDNA change:** c.460+16G>T

- **ClinVar accession:** VCV003607857
 - **Last evaluated:** 2025/02/03 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (splice pipeline) on 2026-06-08T07:50:31.536741Z.

Schema: `reference/card schemaextension.md` (S1–S3). WFS1: UniProt O76024, AlphaFold v6.