

# c.315+18G\_C — WFS1 Molecular Atlas Card

**Variant type:** Splice site

**Boundary:** donor (5' splice site) · intronic offset +18

**Nearest protein position:** ~105 (N-terminal cytoplasmic (intrinsically disordered))

---

## SCHEMA CATEGORY: S3 — MINIMAL PREDICTED SPLICING IMPACT (SPLICEAI $\Delta S$ 0.00)

---

SpliceAI predicts little splicing disruption at this donor (5') site (max  $\Delta S$  0.00 < 0.2; acceptor-gain 0.00, acceptor-loss 0.00, donor-gain 0.00, donor-loss 0.00). The variant may be tolerated or act through a weak/again-tissue-specific mechanism; wet-lab RNA validation is the arbiter before any therapeutic call.

---

## SPLICE PREDICTION

---

- **Affected site:** donor (5' splice site), extended splice region
- **SpliceAI delta scores** (GRCh38 chr4:6287193 G>C):
  - acceptor gain **0.00** · acceptor loss **0.00**
  - donor gain **0.00** · donor loss **0.00**
- **Predicted outcome:** Minimal predicted splicing impact (SpliceAI  $\Delta S$  0.00)

---

## CLINICAL EVIDENCE

---

- **Classification:** Likely benign
- **Review status:** criteria provided, single submitter
- **cDNA change:** c.315+18G>C
- **ClinVar accession:** VCV001625372

- **Last evaluated:** 2025/08/07 00:00
  - **Submissions:** 1
- 

*Card generated by `wolfram-atlas-batch` (splice pipeline) on 2026-06-08T07:49:59.199228Z.*

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