

c.712+131C_T — WFS1 Molecular Atlas Card

Variant type: Splice site

Boundary: donor (5' splice site) · intronic offset +131

Nearest protein position: ~238 (N-terminal cytoplasmic (intrinsically disordered))

SCHEMA CATEGORY: S3 — MINIMAL PREDICTED SPLICING IMPACT (SPliceAI ΔS 0.01)

SpliceAI predicts little splicing disruption at this donor (5') site (max ΔS 0.01 < 0.2; acceptor-gain 0.00, acceptor-loss 0.01, donor-gain 0.01, 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 chr4:6292128 C>T):
 - acceptor gain **0.00** · acceptor loss **0.01**
 - donor gain **0.01** · donor loss **0.00**
- **Predicted outcome:** Minimal predicted splicing impact (SpliceAI ΔS 0.01)

CLINICAL EVIDENCE

- **Classification:** Benign
- **Review status:** criteria provided, single submitter
- **cDNA change:** c.712+131C>T
- **ClinVar accession:** VCV001222165

- **Last evaluated:** 2018/06/23 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (splice pipeline) on 2026-06-08T07:51:46.924660Z.

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