

# c.712+114A\_G — WFS1 Molecular Atlas Card

**Variant type:** Splice site

**Boundary:** donor (5' splice site) · intronic offset +114

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

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## SCHEMA CATEGORY: S3 — MINIMAL PREDICTED SPLICING IMPACT (SPICEAI $\Delta$ S 0.19)

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SpliceAI predicts little splicing disruption at this donor (5') site (max  $\Delta$ S 0.19 < 0.2; acceptor-gain 0.19, acceptor-loss 0.03, 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.

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## SPLICE PREDICTION

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- **Affected site:** donor (5' splice site), extended splice region
- **SpliceAI delta scores** (GRCh38 chr4:6292111 A>G):
  - acceptor gain **0.19** · acceptor loss **0.03**
  - donor gain **0.00** · donor loss **0.00**
- **Predicted outcome:** Minimal predicted splicing impact (SpliceAI  $\Delta$ S 0.19)

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## CLINICAL EVIDENCE

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- **Classification:** Benign
- **Review status:** criteria provided, single submitter
- **cDNA change:** c.712+114A>G
- **ClinVar accession:** VCV001226503

- **Last evaluated:** 2018/06/23 00:00
  - **Submissions:** 1
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*Card generated by `wolfram-atlas-batch` (splice pipeline) on 2026-06-08T07:51:43.331810Z.*

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