RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · SPLICE

c.712+16G_A — WFS1 Molecular Atlas Card

Variant type: Splice site

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

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 0.00, 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 chr4:6292013 G>A):

- acceptor gain **0.00** · acceptor loss **0.00**

- donor gain **0.01** · donor loss **0.00**

- **Predicted outcome:** Minimal predicted splicing impact (SpliceAI ΔS 0.01)

CLINICAL EVIDENCE

- **Classification:** Benign/Likely benign
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Wolfram syndrome 1
- **cDNA change:** c.712+16G>A

- **ClinVar accession:** VCV000215380
- **Last evaluated:** 2026/03/01 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (splice pipeline) on 2026-06-08T07:51:50.505307Z.

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