RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · SPLICE

c.862-10T_G — WFS1 Molecular Atlas Card

Variant type: Splice site

Boundary: acceptor (3' splice site) · intronic offset -10

Nearest protein position: ~288 (N-terminal cytoplasmic (intrinsically disordered))

SCHEMA CATEGORY: S3 — MINIMAL PREDICTED SPLICING IMPACT (SPLICEAI ΔS 0.02)

SpliceAI predicts little splicing disruption at this acceptor (3') site (max ΔS 0.02 < 0.2; acceptor-gain 0.00, acceptor-loss 0.02, 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:** acceptor (3' splice site), extended splice region
- **SpliceAI delta scores** (GRCh38 chr4:6300647 T>G):

\- acceptor gain **0.00** · acceptor loss **0.02**

\- donor gain **0.00** · donor loss **0.00**

- **Predicted outcome:** Minimal predicted splicing impact (SpliceAI ΔS 0.02)

CLINICAL EVIDENCE

- **Classification:** Likely benign
- **Review status:** criteria provided, single submitter
- **cDNA change:** c.862-10T>G
- **ClinVar accession:** VCV001628873

- **Last evaluated:** 2021/12/22 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (splice pipeline) on 2026-06-08T07:53:34.994532Z.

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