GENE*ie* Report: NM_001142784.3:c.331+6delT

This report was produced for **Demo Admin** of **Demo Account** on Monday, 2 September 2024

This report is for research use only. It is not intended for diagnostic or clinical purposes.

Splice-Neutral

99% PROBABILITY

- Annotated donor splice-site variant.
- Two lines of concordant evidence.

NIF	Similar Impact	Same Sequence	SpliceVault Top4	Cryptic Risk
★★★	☆☆☆	★★☆	N/A	No

Variant	NM_001142784.3:c.331+6delT		
Gene	*IL1RA*	**Coordinates (GRCh38)**	9:34656913 GT>G
Location	(NM_001142784.3) Intron 4 donor / Distance from donor: +6		
Variant Databases	Variant not previously reported in HGMD, ClinVar or LOVD		

Native Intron Frequency (NIF)

Relative frequency of a given splice-site sequence among all human splice-site sequences

Zone	Window	Reference (REF)	Variant (VAR)	Frequency		% Scale		NIF Impact VAR/REF
				REF	VAR	REF	VAR	
U5	E4 - D5	GGCT**gt**gagttg	GGCT**gt**gagtgg	124	124	0.26	0.26	0%
U1	E3 - D6	GGCT**gt**gagttg	GGCT**gt**gagtgg	503	503	0.48	0.59	+23%
	E2 - D7	GGCT**gt**gagttg	GGCT**gt**gagtgg	304	601			
U6	E1 - D8	GGCT**gt**gagttg	GGCT**gt**gagtgg	227	375	0.47	0.58	+23%

NIF-ML Power (Machine Learning)	
ML Score	0.00
Pathogenic	0%
Benign	100%
n	37

Cryptic Splice-site Modification

Cryptic splice-site overlapping the annotated splice-site that is modified by this variant

Splice-site Position	Reference (REF)	Variant (VAR)	REF	VAR	SpliceVault Top-10	In Frame
			U5 | U1 | U6	U5 | U1 | U6		
Annotated	GGCT**gt**gagttg	GGCT**gt**gagtgg	0.26 | 0.48 | 0.47	0.26 | 0.59 | 0.58	N/A	N/A
+5	gtga**gt**tgggga	gtga**gt**ggggag	0.01 | 0.01 | 0.00	0.02 | 0.00 | 0.00	Y	N

Same Splice-site Variants

Variants affecting the same splice-site as the test variant

Test Variant	Distance from Donor	NIF Impact	Classification	Variant ID
		U5 | U1 | U6		
NM_001142784.3:c.331+6delT	+6	0% | +23% | +23%	Not reported	Not reported

Similar Impact Variants	Distance from Donor	NIF Impact	Classification	Variant ID
		U5 | U1 | U6		
No Similar Impact variants reported at this splice-site				

Different Impact Variants	Distance from Donor	NIF Impact	Classification	Variant ID
		U5 | U1 | U6		
No Different Impact variants reported at this splice-site				

Same Sequence Variants

Variants making the same sequence change at a different splice-site

Zone	Window	Reference (REF)	Variant (VAR)	NIF Impact	Pathogenic		Benign	
				VAR/REF	introns	*n*	introns	*n*
U5	E4 - D5	GGCT**gt**gagttg	GGCT**gt**gagtgg	0%	-	-	-	-
U1	E3 - D6	GGCT**gt**gagttg	GGCT**gt**gagtgg	+23%	0	0	2	2
	E2 - D7	GGCT**gt**gagttg	GGCT**gt**gagtgg		0	0	0	0
U6	E1 - D8	GGCT**gt**gagttg	GGCT**gt**gagtgg	+23%	0	0	0	0

E4 - D5	NIF Impact	Classification	Variant ID
	U5 | U1 | U6		
This test variant does not affect this window			

E3 - D6	NIF Impact	Classification	Variant ID
	U5 | U1 | U6		
NM_015627.3(LDLRAP1):c.616+7C>A	0% | +21% | +20%	LB	VCV001130303
NM_000527.5(LDLR):c.190+7C>A	0% | +21% | +20%	LB	VCV001552715

E2 - D7	NIF Impact	Classification	Variant ID
	U5 | U1 | U6		
No same sequence variants for this window			

E1 - D8	NIF Impact	Classification	Variant ID
	U5 | U1 | U6		
No same sequence variants for this window			

SpliceVault Top-10

Natural splicing mistakes around this splice-site that may be enhanced by this variant

Event	Same Frame	Skipped Exons	Cryptic Position	Cryptic mean NIF	Samples (GTEx)	Samples (SRA)	Max Reads (GTEx)	Total Samples	Splice Junction
Annotated Splicing	✓				19,058	144,599	610	163,657	34656909-34657034
Exon Skipping		4			8,767	25,268	21	34,035	34655666-34657034
Exon Skipping	✓	3-4			2,138	7,172	8	9,310	34655318-34657034
Exon Skipping	✓	4-5			233	549	5	782	34655666-34657302
Cryptic donor	✓		-36	0.03	129	273	4	402	34656873-34657034
Cryptic donor [1]			+5	0.01	1	1	1	2	34656913-34657034
Cryptic donor			-19	0.01	0	1	0	1	34656890-34657034

[1] Splice-site is modified by this variant (see Cryptic Splice Site Modification table above)

Understanding your report

Assertion and Probability

Splice - Altering

Clinical and genetic evidence indicate this variant has a given probability of altering pre-mRNA splicing for this gene.

Splice-Neutral

Clinical and genetic evidence indicate this variant has a given probability of maintaining the existing pattern of pre-mRNA splicing for this gene

Uncertain

Clinical and genetic evidence for pre-mRNA splicing disruption by this variant is weak and/or conflicting; and cannot be assigned a probability.

Flags

Summary

Native Intron Frequency (NIF)

NIF-ML Power (Machine Learning)

Cryptic Splice-site Modification

Same Splice-site Variants

Same Sequence Variants

SpliceVault Top-10

Report MetaData

REPORT DATE	GENEIE VERSION	API VERSION
02-Sept-2024	0.0.223.0	0.0.50

ML DONOR VERSION	ML ACCEPTOR VERSION	CLINVAR RELEASE
v1.0.6	v1.0.5	2023-07

HGMD RELEASE
2019.1