Appendix 4. Results of splice sites analysis using the Shannon Human Splicing Pipeline.

Gene Name	*EFTUD2*	*EFTUD2*	*EFTUD2*	*EFTUD2*	*PRPF4*
Chromosome	17	17	17	17	9
Splice site coordinate	42,939,388	42,954,217	42,963,479	42,938,786	116,041,841
Input variant	T/A	C/T	G/C	C/G	T/G
Ri-initial	−13.32	−2.34	2.91	10.68	−2.09
Ri-final	5.3	12.37	6.78	13.05	1.28
ΔRi	18.62	14.71	3.87	2.37	3.38
Type	DONOR	ACCEPTOR	DONOR	ACCEPTOR	DONOR
Loc. of nearest nat. site	42,940,080	42,953,469	42,963,952	42,937,912	116,041,412
Ri of nearest nat. site	3.21	10.16	4.74	3.3	−1.45
Cryptic Ri relative to nat.site	GREATER	GREATER	GREATER	GREATER	GREATER
Carriers	1	1	1	1	1
Variant coverage		1 out of 4			1 out of 4
Observations	Not concordant with NNSPLICE	False positive	Not concordant with NNSPLICE	Not concordant with NNSPLICE	False positive

Ri is a measure of the likelihood of a sequence to be used as acceptor or donor splice site. ΔRi is the difference between the R value of a genomic coordinate in the presence and in the absence of the nucleotide variation. Nat.site is the closest natural splice site.