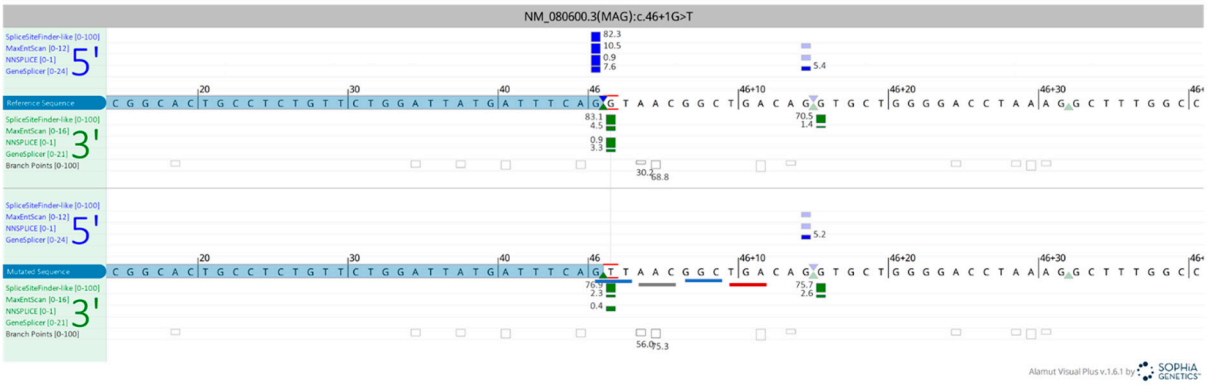
(A)



(B)



(C)

Features	Values	Descriptions
dbscSNV ADA:	1.00	*Raw score 0-1, threshold ≥ 0.8 for impact*
dbscSNV RF:	0.92	*Raw score 0-1, threshold ≥ 0.8 for impact*
spliceAI AG:	0.00 (-47)	*Acceptor Gain, Δ score 0-1 (relative position in bp), thresholds ≥ 0.2|0.5|0.8 for impact*
spliceAI AL:	0.21 (0)	*Acceptor Loss, Δ score 0-1 (relative position in bp), thresholds ≥ 0.2|0.5|0.8 for impact*
spliceAI DG:	0.32 (48)	*Donor Gain, Δ score 0-1 (relative position in bp), thresholds ≥ 0.2|0.5|0.8 for impact*
spliceAI DL:	0.92 (-1)	*Donor Loss, Δ score 0-1 (relative position in bp), thresholds ≥ 0.2|0.5|0.8 for impact*
AbSplice Max Score:	0.31 (Brain Amygdala)	*AbSplice Max Tissue score, thresholds ≥ 0.01|0.05|0.2 for impact*

Figure S1: *In silico* prediction of the *MAG*: c.46+1G>T variant. (**A**) *In silico* splice prediction of the wild-type (top track, red marked G) and c.46+1G>T (bottom track, red marked T). Only scores that are predicted to change are shown. (A) The *MAG* c.46+1G>T variant occurs in the first coding exon. (**B**) SpliceAI Visual prediction. The mutant base (T, bottom track) and wild-type (G, top track) are indicated by pink and green bars, respectively, to the right of the exon border. **(C)** Splice prediction scores including AbSplice, dbscSNV and Splice AI scores [score range: 0–1]. Abbreviations: Ref, reference; Mut, mutant; WT, wild-type.

Table S1: Oligos designed for Sanger sequencing

***MAG*:** ENST00000361922.8:c.46+1G>T	Left primer	cgggcaatgaaacaagaccc
	Right primer	agctcatccgggaagtcaaa

Table S2. List of primers used for the minigene assay

Primer name	**Primer sequence (5´-3´)**	**Purpose**
hu_MAG_Ex3 XhoI_F	aattctcgagCGGGCAATGAAACAAGACCC	Amplification of gDNA and cloning
hu_MAG_Ex3 BamHI_R	attggatccCGAGATGGACGAGGGCATC	Amplification of gDNA and cloning
SD6 F	TCTGAGTCACCTGGACAACC	Colony PCR and RT-PCR
SA2 R	ATCTCAGTGGTATTTGTGAGC	RT-PCR