

Supplementary Figure 2: c. 1214+5G>C – splice analysis

	Exon 9	← Intron 9 →	Exon 10
Wild type	CAA gtaa g gtgc -----		cacttggcag GTG
MaxEntScan (log odds)	10.08	wild-type Enhancer motif	6.97
NNSPLICE 0.9 (0-1)	0.99	SRp55 (value: 74.12 (0-100))	0.78
Human Splice Finder (0-100)	89.42		90.75
Mutated	CAA gtaa c gtgc -----		cacttggcag GTG
MaxEntScan (log odds)	4.45	mutated Enhancer motif	6.97
NNSPLICE 0.9 (0-1)	0.69	SRp55 (value: 0 site broken)	0.78
Human Splice Finder (0-100)	77.41		90.75
	wild-type donor site: broken		predicted splice acceptor site