Additional file

Table S1. Functional alteration prediction of two splicing mutations in UNC13D gene with HSF3.

Mutation	Predicted signal	Prediction algorithm	cDNA Position	Interpretation
c.2709+1G>A	Broken WT Donor Site	1 - HSF Matrices	CAGCAGg t g a g g c c c	Alteration of the WT donor site,
		2 - MaxEnt	80 82 84 86 88 90 92	most probably affecting splicing.
c.1299-1G>A	Broken WT Acceptor Site	1 - HSF Matrices	acacagetetttecatettgcagGGTCC	Alteration of the WT acceptor site, most probably affecting splicing.
		2 - MaxEnt		
	New Acceptor Site	1 - HSF Matrices	cacagetetttecatettgcaaGGTCCT	Activation of an intronic cryptic
		2 - MaxEnt	-20 -15 -10 -5 0 5	acceptor site. Potential alteration of splicing.