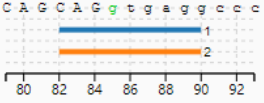
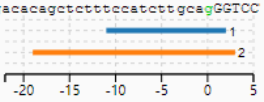
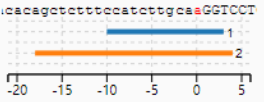


Additional file

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 |  | Alteration of the WT donor site, most probably affecting splicing. |
| | | 2 - MaxEnt | | |
| c.1299-1G>A | Broken WT Acceptor Site | 1 - HSF Matrices |  | Alteration of the WT acceptor site, most probably affecting splicing. |
| | | 2 - MaxEnt | | |
| | New Acceptor Site | 1 - HSF Matrices |  | Activation of an intronic cryptic acceptor site. Potential alteration of splicing. |
| | | 2 - MaxEnt | | |