Supplementary Online Content


eFigure 1. Brain MRI of the RTN4IP1 Index Case of Family 11

eFigure 2. Aberrant RTN4IP1 RNA Splicing Induced by the De Novo c.806+1G>A Mutation

This supplementary material has been provided by the authors to give readers additional information about their work.
**eFigure 1.** Brain MRI of the *RTN4IP1* Index Case of Family 11

MRI scan of the brain showed symmetrical T2 high signal change with associated swelling in the posterolateral aspect (white arrows) of both putamina, suggestive of a mitochondrial disorder.
eFigure 2. Aberrant RTN4IP1 RNA Splicing Induced by the De Novo c.806+1G>A Mutation

Amplification of cDNA across RTN4IP1 exons 4-7 shows an aberrant splicing pattern for RTN4IP1 RNA transcripts in the index patient of Family 11 (F11) with the c.806+1G>A mutation, revealing two shorter abnormal splice products consistent with exon skipping, compared to a control mRNA (Cont.).