

Supplemental Data

A Heterozygous Truncating Mutation in *RRM2B*

Causes Autosomal-Dominant Progressive External

Ophthalmoplegia with Multiple mtDNA Deletions

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Table S1. Primer Sequences Used for Sequencing of the *RRM2B* Gene

Primer Name	Primer Sequence 5' → 3'
<i>RRM2B</i> exon 1 forward	CTTGCCGAGACAGGGATAAT
<i>RRM2B</i> exon 1 reverse	GCGGCGAATAACATTTTCCTA
<i>RRM2B</i> exon 2 forward	ACAGGCTCTCAAACCAATGC
<i>RRM2B</i> exon 2 reverse	TCTCAGTAATTCCAACACTTATCTTC
<i>RRM2B</i> exon 3 forward	TGCAGTGCTAACATCTTTCTTTTT
<i>RRM2B</i> exon 3 reverse	AGACATCTTGTCTTTGGCTGAA
<i>RRM2B</i> exon 4 forward	GCGATAATGCTGATGTCCAG
<i>RRM2B</i> exon 4 reverse	TGCTCAGAAACTTCCATACTTGA
<i>RRM2B</i> exon 5 forward	GGCACCTGTCGTCCTAGC
<i>RRM2B</i> exon 5 reverse	TCCATGCTGTAACTTTGATTCG
<i>RRM2B</i> exon 6 forward	AGGCATACCTTTCTGCTCCA
<i>RRM2B</i> exon 6 reverse	GAGATGGGAGGAATGCTTGA
<i>RRM2B</i> exon 7 forward	AATGCTTGACCCATCACCAT
<i>RRM2B</i> exon 7 reverse	CAGGCTGACCAGAATAAAATTG
<i>RRM2B</i> exon 8 forward	TTTCCTTGGACAAAATCAGTCTT
<i>RRM2B</i> exon 8 reverse	TGCAAAACCTTGTTTCATACAAAA
<i>RRM2B</i> exon 9 forward	CCTGACCTCTGTTTTGGTCA
<i>RRM2B</i> exon 9 reverse	AGCAAACCCCCAGTCCTTTA
<i>RRM2B</i> cDNA (partial) forward	GGCCTCATTGGAATGAATTG
<i>RRM2B</i> cDNA (partial) reverse	GCATCCAAGGTGAAGACGTT

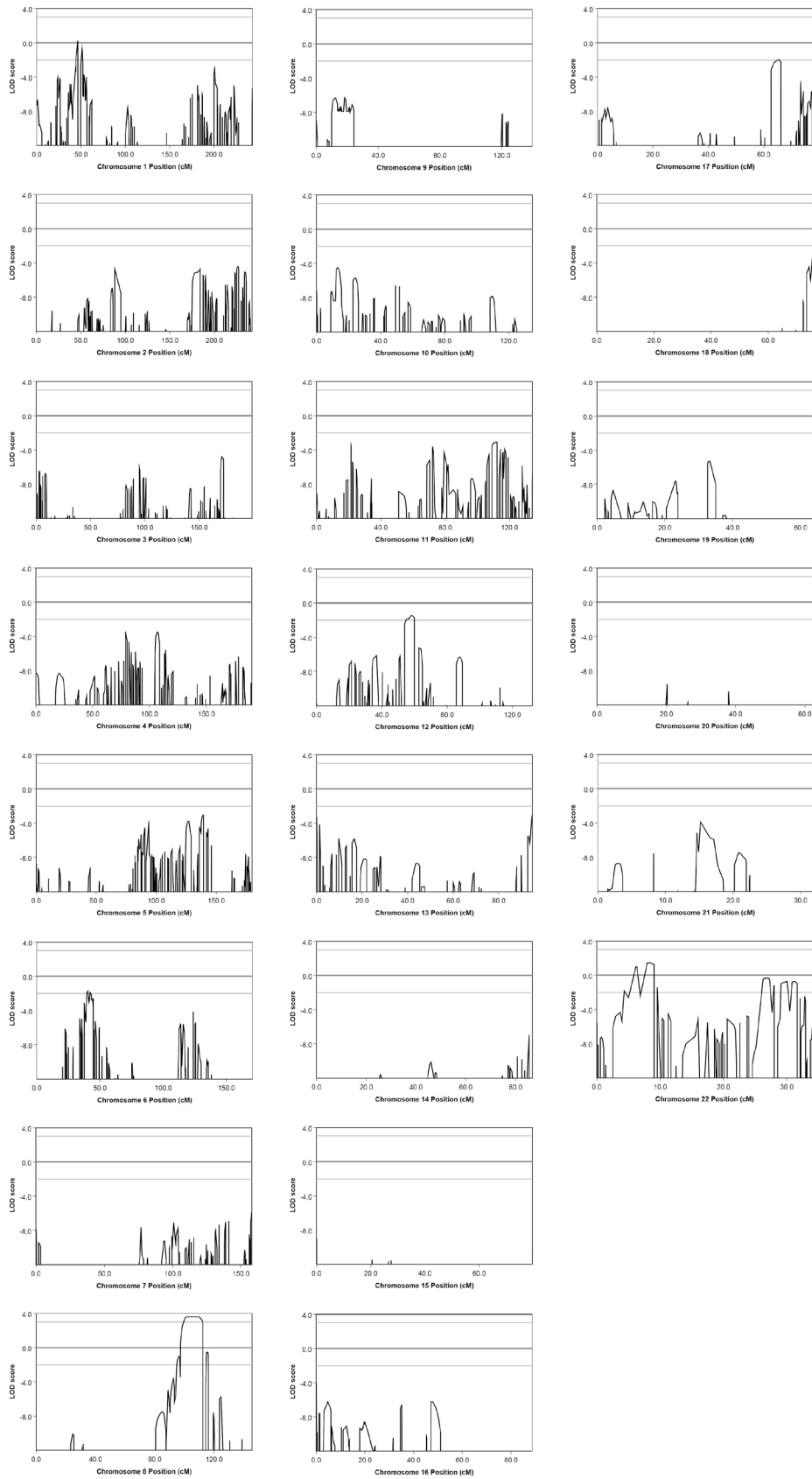


Figure S1. Multipoint LOD Score Analysis Showing Region of Significant Linkage on Chromosome 8