

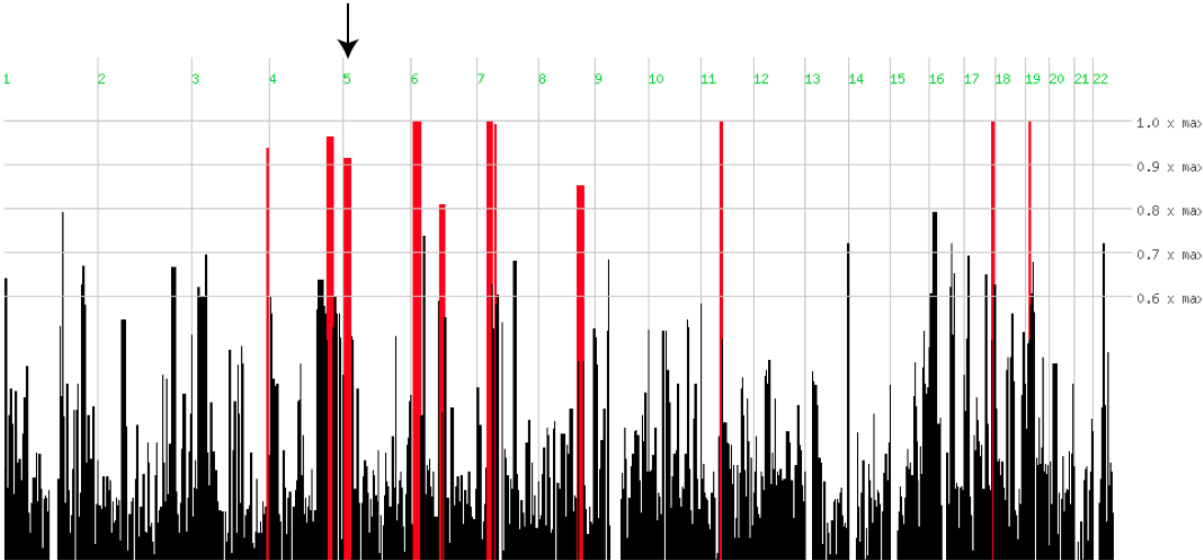
Supplementary Figure Legends

Supplementary Figure 1. Homozygosity plot for Family-1455. Red bars represent stretches of homozygosity in the genome above the threshold of 5 cM. X-axis: chromosomal position. Y-axis: degree of homozygosity. Arrow points to homozygous interval containing the *NSUN2* gene.

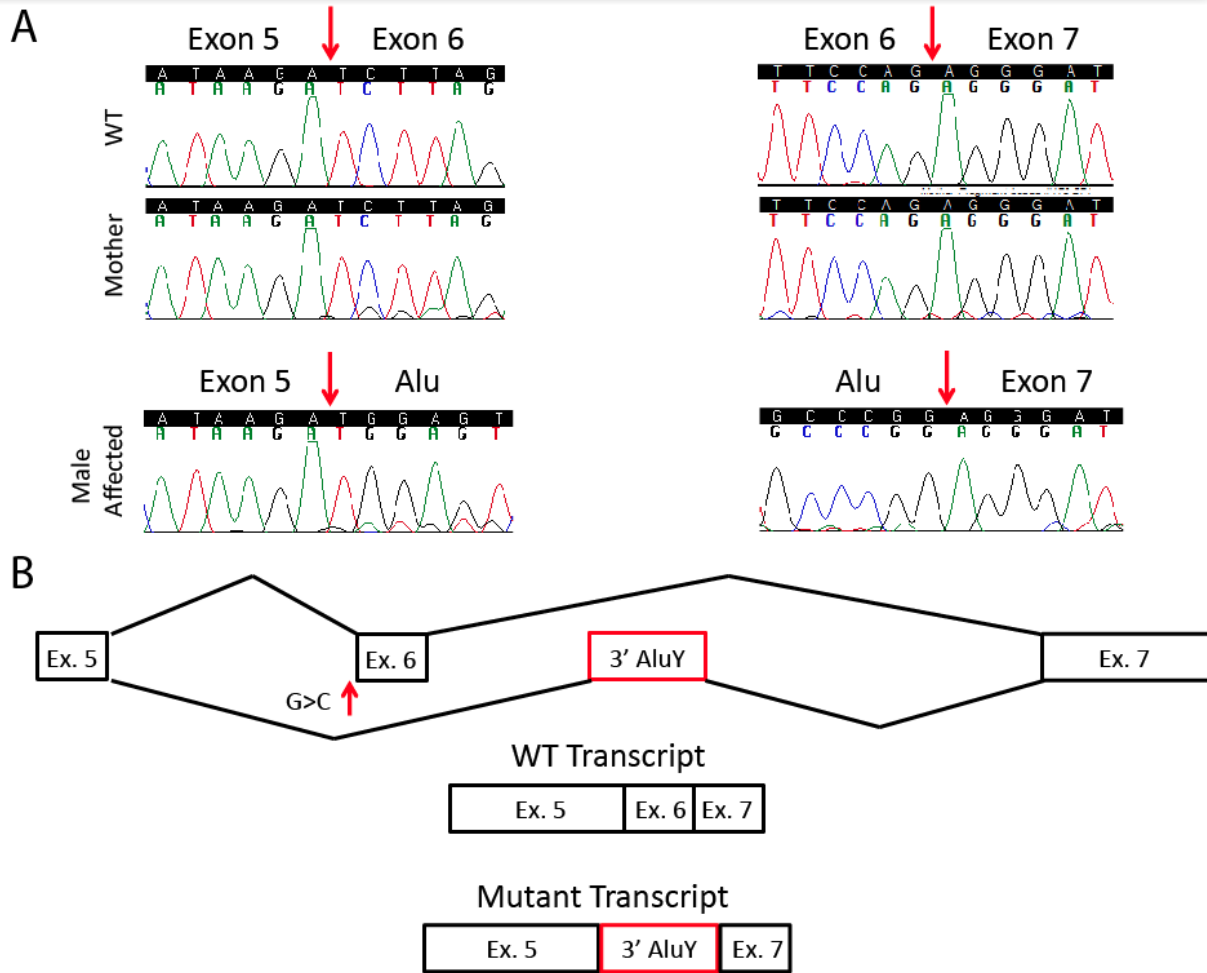
Supplementary Figure 2. Aberrant splicing of *NSUN2* in Family-1455. (A) Sequence chromatogram in control (wt) and mother from 210 bp product compared with 250 bp product from two affected members. Annotation shows splicing from exon 5 to the AluY to Exon 7 accounts for the 250 bp mutant transcript product. (B) Schematic of aberrant splicing in the affected individuals.

Supplementary Figure 3. Predicted *NSUN2* open reading frame (ORF) for control and affected. Blue: Exon 6; Red: 3' AluY arm.

Supplementary Figure 1



Supplementary Figure 2



Supplemental Fig. 3

Wildtype ORF

ATGGGGCGGCGGTTCGCGGGGTTCGGCGGCTCCAGCAACAGCAGCGGCCGGAGGACGCGGAGGATGGCGCCG
AGGGTGGTGGAAAGCGCGGCGAGGCGGGCTGGGAAGGAGGCTACCCCGAGATCGTCAAGGAGAACAAGCT
GTTTCGAGCACTACTACCAGGAGCTCAAGATCGTGCCCGAGGGCGAGTGGGGCCAGTTCATGGACGCTCTC
AGGGAGCCGCTCCCGGCCACTTTAAGAATTACTGGTTACAAAAGCCACGCAAAAAGAGATTCTCCATTGCT
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ACTGAGTTGGTATCCTGAAGAACTTGCCTGGCACACAAATTTAAGTCGAAAAATCTTGAGAAAAATCGCCA
CACTTGAAAAGTTTCATCAGTTTCTAGTTAGTGAAACAGAATCTGGAAATATTAGTCGTCAAGAAGCTG
TTAGCATGATCCCACCACTGCTCCTCAACGTGCGGCCTCATCATAAG

ATCTTAGATATGTGTGCAGCACCTGGCTCAAAGACCACACAGTTAATTGAAATGCTACATGCCGACATGA
ATGTCCCCTTTCCAG

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GA

Mutant ORF

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ACTGAGTTGGTATCCTGAAGAAGTTGCCTGGCACAACAATTTAAGTCGAAAAATCTTGAGAAAAATCGCCA
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GG

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