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Supplemental Data

Mutations of *POLR3A* Encoding a Catalytic Subunit

of RNA Polymerase Pol III Cause a Recessive

Hypomyelinating Leukodystrophy

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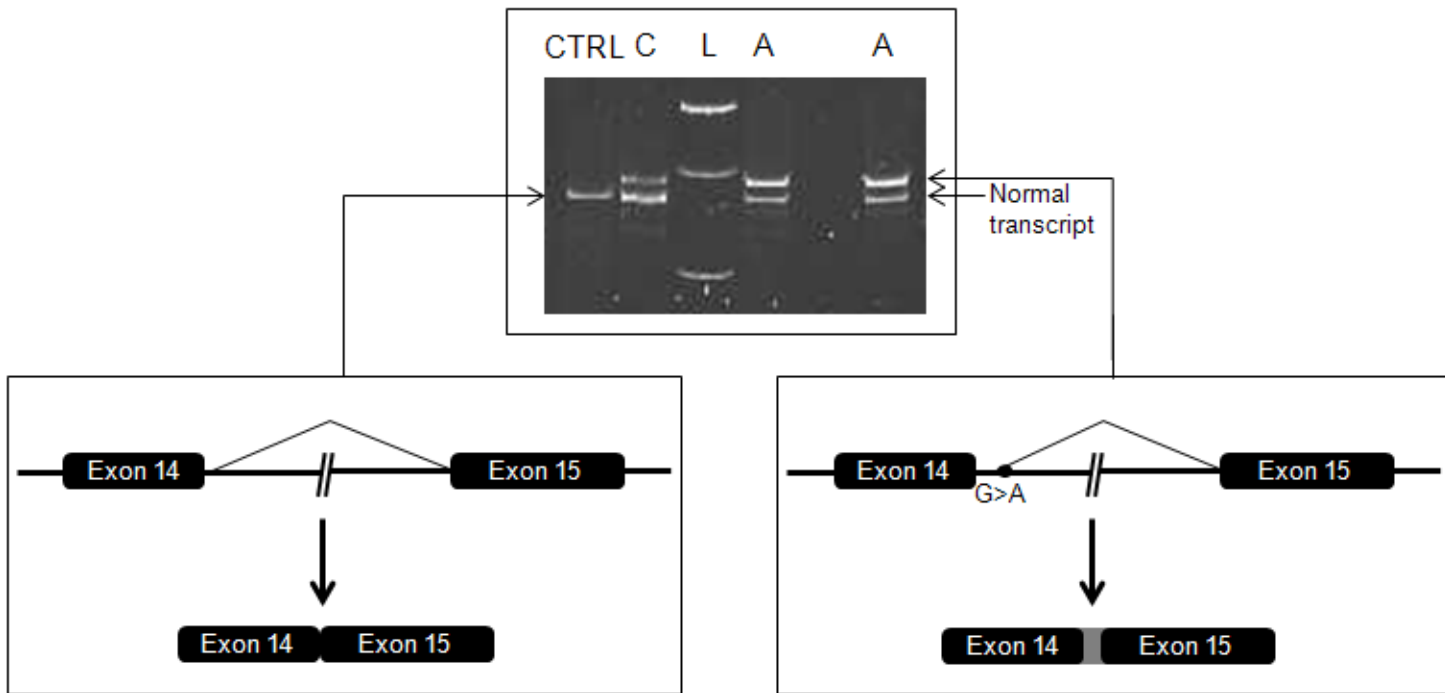


Figure S1. Splice Site Mutation c.2003+18G>A

Legend: Agarose gel of the amplification of cDNA of a control (CTRL), a carrier (C) and two affected individuals (A) homozygous for the splice site mutation c.2003+18G>A. The G to A substitution creates a new donor splice site, causing the retention of 19 nucleotides from intron 14. In the homozygous participants, cDNA amplification revealed that both the normal allele and the allele with aberrant splicing were produced. These results were proven by cDNA sequencing. A: Affected, C: Carrier, CTRL: Control, L: Ladder.

Table S1. POLR3A Gene Primers

NAME	EXON(S)	PRIMER	PRODUCT SIZE (bp)
POLR3A_redo-exon1-F	1	CTTCCAGTAAGCGCCACCA	464
POLR3A_redo-exon1-R		ACCAACGCTGGTAGTTCTGG	
POLR3A_exon2-3-F	2 - 3	GGTAGGAAAAGCCAGTGGAG	890
POLR3A_exon2-3-R		CTTGAGTGATTTAACGTAGTGTGAG	
POLR3A_exon4-F	4	TGGTCACGTGGTTAAGGGTA	374
POLR3A_exon4-R		GAAAAGCTGACTCCCGAACA	
POLR3A_exon4-5-F	4 - 5	AACACAGGCTTCCTCTCTGC	887
POLR3A_exon4-5-R		CTGGACCTCTCATTTTGGAAAG	
POLR3A_exon5-F	5	GGGAAACGGACCTCTCATCT	399
POLR3A_exon5-R		TTTTGGAAGAAAGTGGGTGTC	
POLR3A_exon6-7-F	6 - 7	CAATTTTAAGGCTCTTGCAGC	822
POLR3A_exon6-7-R		TGTGCTCATGGGAACAATAAG	
POLR3A_exon8-F	8	ACAAGATGTGCAGAAGCCAG	425
POLR3A_exon8-R		AACCAAACGTGGGTTGAGTG	
POLR3A_redo-1exon8-F	8	CCGTTCTTATTGTTCCCATGA	299
POLR3A_redo-1exon8-R		GGGGACTTTCTACTGCCTGTT	
POLR3A_redo-2exon8-F	8	CAGGCTTTCCTCAGTCTCTCC	299
POLR3A_redo-2exon8-R		TTGTTGCAATACCAAATAACG	
POLR3A_exon9-F	9	CCAGGATGCCTCTCTTTCTC	387
POLR3A_exon9-R		AATGACATACCTTGGCTCAG	
POLR3A_redo-2exon10-F	10	TGTTGCCAGTCTGTTCTCA	669
POLR3A_redo-2exon10-R		CCGTGGTCCCAGCTACTTT	
POLR3A_exon11-F	11	GACTGGGAATGAGTCCTAAATG	439
POLR3A_exon11-R		TTTAAGAACAGAAAGAGCCTGG	
POLR3A_exon12-F	12	TCATTTCTCAGCCTCGTCTT	295
POLR3A_exon12-R		TTAAATAGCTTTTCAAGTCATCACA	
POLR3A_exon12-13-F	12 - 13	CAGCCTCGTCTTCTGTCTAC	973
POLR3A_exon12-13-R		GAAGCCTGAGAGTCAGTGGG	
POLR3A_exon13-F	13	GCAGCTGTTGTTTCAATTAC	397
POLR3A_exon13-R		GGTCTCAATCCCCCTCAGT	
POLR3A_exon14-F	14	TTGTGTGTTGGTGCACAGG	439
POLR3A_exon14-R		TCCACCTAAGGCTTACAGAAAC	
POLR3A_exon15-F	15	TCCAAGTTTTCTTTAACC	493
POLR3A_exon15-R		GCACACATGATTTAGCCTGG	
POLR3A_exon16-F	16	TGAATTGTGATGAATTCTGTGAAG	473
POLR3A_exon16-R		AACAGGACAGACGCCAATG	
POLR3A_exon17-F	17	TTTACTGATCGGAAATCGGG	412
POLR3A_exon17-R		CTTCAAAGCAGTTAAACTTCCC	
POLR3A_exon18-F	18	TGTTCCAGGACCATGACCTC	416
POLR3A_exon18-R		CATGAGACACTGTGCCCG	
POLR3A_exon19-F	19	GGGCAAAGAAAGCCTGC	438
POLR3A_exon19-R		TTCCAGGGCATCTACCTTTC	
POLR3A_exon20-F	20	TGTTCTTGGGGTGAGAGGAC	469
POLR3A_exon20-R		AAAGATACTAATCCAGTGCTTGGG	
POLR3A_exon21-F	21	AAAATCACAGGTTGCTAAAAGC	383
POLR3A_exon21-R		GGCCATCTTTGAACTTTCTAAC	
POLR3A_exon22-23-F	22-23	GAAGCAGTATCCAGATTGGGTC	554
POLR3A_exon22-23-R		CCCCTACAGAAAATGTGACAG	
POLR3A_exon24-F	24	CCCTTCCGACCCTAATAGC	423
POLR3A_exon24-R		CATGCCACCCAGAGTTTAAG	
POLR3A_exon25-26-F	25-26	GCTCAGTAAACTGGGGTTGC	668
POLR3A_exon25-26-R		TTAGAAAGAAAATGTGAGGGG	
POLR3A_exon27-F	27	GCATATTTATGGTATTAATTCATGGT	497
POLR3A_exon27-R		GGGACTGCACCTCATCTAA	
POLR3A_redo-exon28-F	28	GGCCTTATCCCCAAGGAGT	574
POLR3A_redo-exon28-R		CAGAGCTTGCAGTGAGCA	
POLR3A_redo-2exon28-F	28	AAAGCTGGGGTGATCAAGGT	300
POLR3A_redo-2exon28-R		CAGATGGCACAAGGAAGACA	
POLR3A_exon29-F	29	ATCCATTTCTGGAAAACGGC	420
POLR3A_exon29-R		GCCTGACCAGCATACTTTGG	
POLR3A_exon30-F	30	GCCTTTCACACCTGACGTTG	409
POLR3A_exon30-R		CCAGCAGTTCCAATTCGTG	
POLR3A_exon31_1-F	31	ATGGGAAGGAAAGGGATTTG	978
POLR3A_exon31_1-R		ATGTTACAGAGCTGCCTGGG	
POLR3A_redo-exon31_2-F	31	ACACACTGCTTCCCTGGACT	685
POLR3A_redo-exon31_2-R		GGAAGGGAAGCACTCATTTC	
POLR3A_exon31_3-F	31	GGAAGCCTCACTGGAGTTTG	680
POLR3A_exon31_3-R		CCACACTGATACCCCAAG	

Legend: This Table lists all primers used to sequence the 31 exons of *POLR3A*. Abbreviations used: bp: base pairs, F: forward, R: reverse.