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Supplemental Data

**Expansion of Intronic GGCCTG Hexanucleotide Repeat
in *NOP56* Causes SCA36, a Type of Spinocerebellar Ataxia
Accompanied by Motor Neuron Involvement**

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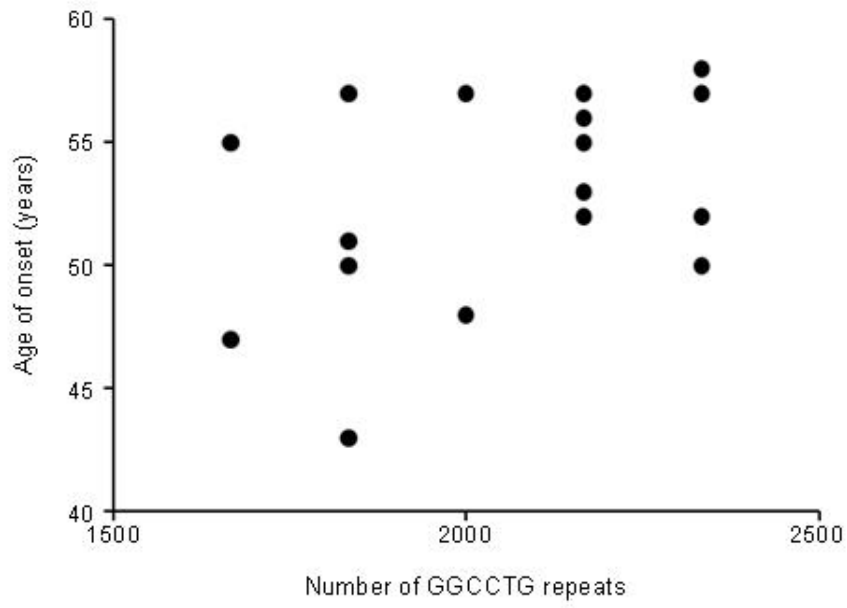


Figure S1. Correlation of the Number of Repeats with Age of Onset

A scatter plot shows no negative correlation between GGCCTG repeat number and onset age (n=17, r=0.42, p=0.09).

Table S1. Primers Used for Amplification of Candidate Genes (Human Build 37.1)

<i>PDYN</i>	exon 3	CTTTGGCCTCTGCTTACCT	TCCAGGCCATCTATAGGGCA
	exon 4	TCCCCTACCTTTATGCAACCA	AACATACTCCCACCGCAGAAGA
<i>STK35</i>	exon 1	CGGATCACGGGAATTTCCG	ATTGGCTGAAAAGTTCGGCT
	exon 2	TGGCTTCCGCTTAAAGGG	CAGGAAAGGAGGGTGTGTCA
	exon 3	GTCCCTTGGAGCAAGTGTGTT	AATCACTTGAAGTTCGGGAGGT
	exon 5	TCTCTTTAGAGCTCTGCCCA	TTGCCACATTGAATTTCTT
<i>TGM3</i>	exon 1	TTATTAATCTGCCCTTCTCC	CTCTGGCTAGCACCTAAAT
	exon 3	AACAGGATGCACAGAGTTC	TCCCTCTGATTTGAGGATGG
	exon 4	TGGCTGTATGTTTGTCCA	TTGGGGCTTGGAGAGATAGAA
	exon 5	TCAGGGGAGGGCTAAAGGT	AGGTGGCCAAATGAAAAGTCTT
	exon 6	TTCTCTGGTCTTCCGCAAT	TGTAAAGAGTGTGGTGCCTA
	exon 7,8	TTCAAATCATGGCTTTGGCT	AATCAACATGGCAAGAGT
	exon 9	TGTTGTATGCTGCACTGTG	TTGTTTAAATCCCATCATGCA
	exon 10	GGTTGCAATGGTCTGGAA	GCAATCCTATCATTCAGGCA
	exon 11	TGAAAGTCGAATGCCTGCTA	TCCAAAGCATTAATACATGGC
	exon 12	AGATCTCCCAACCAAGCTCA	AAAACTCTCTTTGCCCTCTG
	exon 13	TCTCCCTTCTTCAATCCTCA	CCAAACCAAAATGCAAAAGCA
	exon 14	CTCCAACAACAAGGACAGGA	CACTCCCTTTGGCAATTGAA
<i>TGM6</i>	exon 1	TGATTTGTGTCTGTGGGTG	AGTTCATGTGTCTGTGGGA
	exons 2,3	ATGAACAAATGACTGGCCGA	TAAAGTCTTGGCCAGCTCTTG
	exon 4	AAGCCCTCTTGAACCTCT	CCTGGCCAGTGAATGTAAGA
	exon 5	TTGAGGAAGGGTTCCAAGAC	CAGCGAATTAACAAGGGG
	exons 6,7	AAAAACAAGAGTGAAGCCGAT	AGATTCAGGAGAGCTGGCT
	exons 8,9	GATTCACAACATGCAAGCCACA	AAATAAGGCACTTGGCTCAGA
	exon 10	GAGAATCAACAAGGCAATG	AAGGCACTGACCAATGCT
	exons 11,12	TGGCCCTTAGGTTCTTCAAT	ATAGTCTGTGGCTGGTCTCT
	exon 13	ATGTCAAGCCACAAGGTGAA	AGATGAAAGGTGGAGAGGCTC
<i>SNRPB</i>	exon 1	AGCAGCTCTCAATGCGAATTT	CGCCAAAGTCTGTCTT
	exon 2	GTGGCATGGGAAATTCCTA	AGCGTCAATGTCCCAATTT
	exon 3	GGGCTATCTGGGAAAGTTTG	CCTCTTGCAGGGTAACTCTT
	exon 4	AATGGTGTGGCAACATGGA	CTTTTCACTGTCTTACGGGC
	exons 5,6 (<i>SNORD119</i>)	TGGTCTGAGAGTGTAGCAT	GCCCAAGAAATAGCTCAA
	exon 7	ACATAAGAAAGGATGACCT	GGGAAAGAGTAAAGATCAGGG
<i>ZNF343</i>	exons 3,4	GATGGAGATCCGTTGTTGTT	TCAAAGAAAGCCTAATGCTTCA
	exon 5	TGTCAAGGGCTAGAAATGTT	TAGAAGTAAAGCCCAATGGAG
	exon 6	TTTGCAAGGGGAGAGATG	TTATGGCTGCAACTCAATGG
	exon 7-1	GGCATGGCAGTAAAACATTC	TCACAAGGCTTGTCCACA
	exon 7-2	GTGTGGCAAGCTTTAGAA	TACACAGGGTCTACTCCCAT
<i>TMC2</i>	exons 1,2	AAATCTCAACCAAGAAGCCCC	TCCCATGTTAAACACTTGGC
	exon 3	TTTGGGGTGTCTGTCTGA	TTCTGGGATGAAAGAACCCAC
	exon 4	TAGAATTTGGCTCCGCT	AGTCTCCGAGTCTCCATG
	exon 6	TTCTGCCTACCATGCTGTTA	AATTGGCAAGTACGATTTGG
	exon 7	CGTTGGGAAAGTAAAGGTTG	TGGCTGACAAAGTCAAAAC
	exon 8	TTCACTTTCTGACTGTGGCA	CAATTACTTTGACACCAAGCC
	exon 10	AGCTGAGACTGTCTATTGCA	TGCAAGACATGGTTTGA
	exon 11	AGGAAGTGAAGGGAAGAAACA	ATGGCTGGTCAATGTCTTT
	exon 12	TGTCTGGCTATCCTCAAAGC	ACCAAGCAGAACATTTCTTA
	exon 13	ACCAAGTGTGACATGGTGA	GTTGTAAAAGCCAGTCCCAA
	exon 14	CATGTTGATGCTTTGCCA	AACAATGGTCACTTTGGTTC
	exon 15	TGAAAGCCTGAGAAAGGAAAT	CAAGAGATTTGAGTTCCCA
	exon 16	TGCCTCTCTTCAACACAAA	AGGAGCTCAGCAACTTGT
	exon 17	TGCAAGGCTGAAAGATAGA	CAGAAACTGTCTGCTTCTCA
	exon 18	ATGGTCTGGCTGATGATCG	ATTAAGTGGCAATGTGGTG
	exon 20	TCTGGTCTTCCAGGAAAGCA	CCATGCAATTTCTTTCCCT
	exon 21	CAGGACCTTCTCCACATGGA	TGCATGCTGTGATCTTCA
	exon 22	AAATCGCTTGAAGCCAGGA	ATTCACCTGCCAACTTCAT
	exon 23	AGTGAATCCGACAGCTCTCA	TCAAGCGATCCACCCACTT
<i>NOP56</i>	exons 1,2 (<i>MIR1292</i>)	TTCCCAAGTCTTTGCC	ATCTAGAGCTTTCCAAGCCC
	exons 3,4	TGAAGGAAGTGGAGGATCA	CCTTAGGCTCTGTGAAGACAA
	exons 6,7	TGATGGGAGGGATCTAGGTA	AACACAGCTCTGGTAAAGCA
	exon 9-1	TGGATCTTTGCCAATTTCC	TGGTCAAGCATCAGGTGA
	exon 9-2 (<i>SNORD86</i> , <i>SNORD56</i>)	ATGCTGGCAGCCTCAACAA	CAGACAGTCACTCACTCCA
	exon 9-3 (<i>SNORD57</i>)	GGAGAGCTTCGAGAAACAAGT	AAAACAACCCCACTGCTG
	exon 9-4	TGGCTGAGGTAATTTCTCAT	ACTGAGGCTGTCAATGCTGC
<i>SNORD110</i>	exon 1	TCTGCTTTCTGTTCGATGG	TCAGGGGAAAGACACAGTTC
<i>SNORA51</i>	exon 1	CCACCCATAACTGGAGCCT	TGCAAGAGCCACAGTCACT
<i>IDH3B</i>	exons 1,2	AAAAGGAGAAACAGGGCTGA	ACCGATCTGGGATGAGAA
	exons 3,4	AATCTGGCTGGGCTCTCTCT	TGGTTCCTGGAGTAAATA
	exons 5,6	TTACTGATGGGGAATGGGA	TCACAAGCAGTCAAACTGGT
	exons 7,8,9	CCCCAAATCAAAATTTGAGAC	AGATGAAAGAACGCCCTGAGA
	exon 12	ATCTGGCTCTCTTCCATT	AAAGGCGGTTGGCAAGA
<i>EBF4</i>	exon 1,2	GGAAATCGGGAGTACAGTCA	TCAGAAATCTACCGGGGCA
	exon 3	TCCAACATTCAGGCTATCA	TTGATGTTTCAAGGGAGTCA
	exon 4	TTTTTGGCCAAACTCTTGGC	GGACAAGATGGGAGGATGCT
	exon 5,6	AAGTGGGGTATGAGGAAAGGG	TAGACAAGAGGGCAAAAGCCA
	exon 7	TAGGCTTGGAGATGCCA	TAAACAAGCCAGGCTAATGG
	exon 8	ACATCAGCACCTCAGCTCA	TCCAGAAAGTGGCCACT
	exon 9	CCATGATGGGATATGGGAT	ACAAGTGGAGAAAGGAGTGGC
	exon 10,11,12	TTTTTTGACGCGCTGGGA	AAATGTTCAAGGTTGAGATGG
	exon 13,14	GAGTTTTCGGGGGACTTG	TGCTGAAAGGCTGTGATGC
	exon 15,16	AGFAACCAAGTATGGCGCTC	CGGCAAGAAAGGCTAAAAGT
	exon 17	AACAAAGTACCCCAAGGTGCA	AAGAGCAGGTTAGCCAGCAT
	exon 18	AAAGTCTATATCCCTGCC	AGGCTGAGCAGGATGAA
<i>CPXM1</i>	exon 1	TCTGTGTGGTCACTTATG	TGTGTGATGTGTGAGTGC
	exon 2	TGCTGTGGCTCACATGTC	TAAAGTGTCTCTCGCTA
	exon 3	AACTTAAAGCTCACTTCCCA	GACACAGGCACTGGTGTCA
	exon 4,5	ATGGTCTCAGGGTGGGAAGG	AAAGGCAAGAGTGTGTTGGA
	exon 6	TCTAGCTGAGCCCACTAGGGT	AAAGGGTGTTCACAGTGA
	exon 7	AGTCAAGCCAGGGTGGT	TGCACTCTGGTGTGTTCA
	exon 8,9	ACTCTGTCTTCTCGCTGTG	GAACAGACCCAGCACTGTA
	exon 10	TGAAGTGTCCCTCAGAGAAAG	CCTGTGTCTTCCAGAGCAAT
	exon 11,12	TTCAATGTTCCATGGAGCTCA	TGGTACTGTAGCAGAGCTG
<i>C20orf141</i>	exon 1	CACCAAGCTGCTATGAGTCA	TGCTGCCACTTACCTATGGA
	exon 2	TGGCAGGTGGCATTGTA	TTGGCTCCCTGGTATGAT
<i>FAM113A</i>	exon 2	TCACTCTCTCTTATGCAAT	ACTGGACGGAACAGACCAAA
	exon 3	TCTGTCCGTCAGAGGTTT	TTGACAGGCACTATCTGT
	exon 4-1	ATCAACTCTGCTCTGGGAT	TTTTCACTCTCACACCCAT
	exon 4-2	CGGAGCAATATCTTGTCCA	TCAAAGGCTAAAGCCATCA
	exon 4-3	TGTAACAATTTGATGCGGGA	TTGTTTAAAGTAGGCTTGGGA
	exon 4-4	AAAGGACACACTTTGGGCTTG	TCCAGGTGAGCTTCTGTGTT
	exon 4-5	TCTTGTGCCCTAGCACT	GGAGGCAACTTCAATGATT
<i>VPS16</i>	exon 1	AAGTGAAGCTGCCACAGT	TGTGGCTAAGTGGCAGA
	exon 2,3,4	AGCCTTGGGAGACAAATGGA	CGGACCAAACTCAAGTGA
	exon 5,6,7,8	GACACTTCAACATGGCAATGTA	CGAATCAAGGAAGTGTGTC
	exon 9,10,11	GCTGTCCGAGACAAAGGATTA	TGGAAGCAGTAGTGTCTTCA
	exon 12	TGGGTTACTATTGGGAGGATCT	TGGAAGTAAAGCCCGCTT
	exon 13,14	TATAGCCAGTATCCCTGTGACG	TGTTGGGTTACAGGCAATGA
	exon 15,16,17,18	TAAAGGCTTGCAGGAGTGA	AAACAGGAGGCTGATTCCT
	exon 19,20,21	ATCCCTTCAAGGACATCAGATGG	AGCTGACAGGAGCATGAA
	exon 22,23	GGGGTGGGGATATATGTAAT	GGAAACAAGGAGTTTGTGT

<i>PTPRA</i>	exons 1,2,3,4	ATCCAGATGTTTGTGACACCC	ACAGTGGAGCAGATGGAGT	
	exons 5,6,7	AGCCATCCCTCTAGGACATCA	TGCGTCCCAACAAATGTGTAT	
	exons 8,9	TGGTTTAGGTGATTTCTGCC	GCTTCTCTGGTAACTGTGGGA	
	exon 12	TGCGTGGCTACTTTTGTGGA	ATGCCACCAATCTGGCTAAT	
	exon 15	TGAGGAGCATGCATATCAGG	CAATGCTGACATCAATCC	
	exon 16	TGTTGAGGGGATTTGGTCT	CTTCACTATGCTAACCCAAA	
	exon 17	CCAGACCACTGTCCAAAGTIT	AGGGGAAAAACAACACAAAGA	
	exon 20-1	CACCTCAATAAGCCCTGGCAT	TGGGCTTGGACAGATGGAA	
	exon 20-2	TTCCAAGTGTCCAAAGGTA	TGGAAGCTAAAACGGGGTCTA	
	exon 21	TCTTACAGGCTTGGTCCATGA	GGTGAAGCAAACTCACTTCA	
	exon 24	TAAAGGAGCTTGTGGCTGTTT	ACCTTGGCTTCCAAAGTCT	
	exon 25	TGGCATCTTTATACAAAGGTG	ACATGGGAAACCAATGGGAA	
	exon 27	CCTGGCTGGATTTCTTATT	CAAGGACAGAGGGGCTTATTA	
	exon 28	CCTGGCACATACATGGTAA	TCTAGGCACACACTGAGGTT	
	exon 29	AAAGAGTCAAAGGGCTTCT	TTTCCACAGTCTTGGTCA	
	exon 30	TTTTAGTTCTGACCTGCCA	TAATCTTGGAGGACTGCCCTA	
exon 31	TTCTAGCTGGAGGTCAAGATT	TTGGGGCTAGGGTACAGATT		
exon 32	TGCAATTCAGTCCACTTCA	AGTGTTCACAGTGCAGGAT		
exons 33,34	CCAAGTGAACATATGGGAA	AGTGGTGGTGAATCAAT		
exon 35	CAGAAAGCAACCAAGTGTCA	TTGGGAAAGCTAATGACTGC		
exons 36,37	ACGAAAGCTATCAGCGTAA	TAGCAATCCAACTCTGTCTGG		
exon 38	TGAGTCCCTCCACAGCAT	TATGCTCCCAATGCTGCCAT		
exon 39	CAGAGCTCAGGTGAAAGTCA	TTTCTCGGCTGAGGATTTCA		
<i>GNRH2</i>	exon 2,3	GCAGAGAGGGAAAGGGATA	TGAGAAATGGCTGGGGT	
	exon 4	TAGCTGGATCCCTCAAGCTT	GGGGCAATCCCTTATGTTACT	
<i>MRPS26</i>	exon 1	TTCCGTTCCAGAGGCCACA	TTCTCTGCACTCGGACA	
	exon 2,3	TTACAGCACTACCCGCCAGA	TTTGGCTGACTGGCACT	
	exon 4	AGAGCAAGGACTGCTTTCTCA	TGCGTTTGGAAAGTTCTGAC	
<i>OXT</i>	exon 1	AATGAAAGAGGAAAGCCCGTA	TCAAAAATCCGCTCAGCTCT	
	exon 2,3	GGAGCTGAGCGGATTTTGA	AGAAACAGCAACCCGCTCTGT	
<i>AVP</i>	exon 1	TGTCCTCCAGATGCTGAAT	ATGCCATGCTCCCTCT	
	exon 2,3	AAACCAAGTGCAGGAGAT	TCCCACTCTCTCCCTTTC	
<i>UBOX5</i>	exon 4	AAGGAAAGTCAAGTGTGGACG	TGATTTAGAGGTGAGCCTGC	
	exon 5-1	CCTGATCTGGGACAATTCAAGT	TGCGGTTACACTTCTCCAAGT	
	exon 5-2	AGAAGCTGCCGAGATCAATT	TCTGAAAGCAAAAGCTGAGGGG	
	exon 6	TACTCCCGGTGTTCTGTGAT	AGGGTGGGTGTGGAACTGA	
	exon 7	CCACTCCCTACCTGATCAGA	AGCCGAAAGCAATGTGCTAT	
<i>FASTKD5</i>	exon 2-1	TCATTTGTGATCCCTGGCTC	ATGCTGCTTCCAGGCAAA	
	exon 2-2	CCGTGTTCAAGCTATAATGC	ATGTGATCCACGTGAGTAA	
	exon 2-3	AGGTGGTACCACTCTGTTGG	ATACCAAGCAATTCAGACCC	
	exon 2-4	AGGGTTGTGCAAGTATGCTCA	AACTCTACCAACGGTACCA	
	exon 2-5	TAGCAGATAAATCAAGGGCCA	TAGCTCTAACTGCTTGGAT	
<i>ProSAPI1</i>	exon 1	ATTCTTCACTTGGATGGCT	CACAAACCAACCTCCAAGAA	
	exon 2-1	GGGAACCTCAAGGTGAAAT	AGCTCTGGATGAGTCACTG	
	exon 2-2	TCCAGCAAGAGTGGGTGCT	ATTGATTTTGTCCCTCCG	
	exon 3	TTGAGTCCAGGCAAGGAAAT	AGGGAGAACTGTGTCACA	
<i>DDRKG1</i>	exon 1	GGACATACCGTCTGATAAATTC	TTGGAGTCGAGAGAAAGGGGA	
	exon 2	CCTTGCCAGTCAAGCTGAGA	AACAAATGCCAGTCCCAA	
	exons 3,4	AGTGAATTTGCAAGTGGGT	AGGGGACCAAAATAACCAAGG	
	exons 5,6	TTGGGGGAATGGAAGAAATG	GGGTGGAGGGGAGAGAACT	
	exons 7,8	TACAGTGTGTTTCCAGCCACC	TCCTCTTGTAACTGCAATCCA	
	exon 9	TGATTCGACTTCTCAAGAGG	TTATCTAGGTCTTGGGGCA	
	<i>ITPA</i>	exon 1	AGAGAAAGCGGAAAGCAAGG	TTCTTGGCCCAAGCTTTT
		exon 2,3	GTAAGCTTTTGAAGATGGGCA	CGGTCTAGAAAAGTCAACAA
		exon 4	CCAAAGTTAAGAGATTGGCCG	AAAGAAAGGCAATGCTTCTCC
exon 5		TGCTGGGATTATAAGCGGGA	TACAGGGTACGAGCTGCAAGT	
exon 6		CCGCTAACCCAAATTGAGA	TGAAAAGCTGGAAAAGGCTGA	
exon 7		AGCAAAACATTTGCAAGTCT	AGATTCTAGTGTCCACCCCA	
exon 8		ACTCCCTTCTCTGGGGT	TCCACTTCCAGAGTTTCTCA	
<i>SLC4A11</i>		exon 1	AGTCGACGTTTCCAGAAAG	CAGAGCCCTAATGAAACCA
	exon 2,3	TTTTGGACCAACGGCTCTG	AGATAGCGGAGCAAAAGCCA	
	exon 4,5	TTCTCCGCTATGGGATG	TCTTGGAGGCAATGGGAGA	
	exon 6,7	TGATGGCTTCCCTGAGAAAT	TCTTCTCCCAAGTTGGTGG	
	exon 8	TTTTCCCTCCCTAGCAGAGG	CAACATGTTTCTGACACCCCA	
	exon 9,10,11	AAAACCTGCTGCCAGTTCATG	AATGGTGGCCAGAGAAAG	
	exon 12	ATCGCTTTCGGGTCTCTCAA	TTGGGGCAGCAATATGGT	
	exon 13	ACCATAATGCTGCCCAA	TTGATCAAGGGCACACACT	
	exon 14,15,16	TTGATCAAGGGCACACACT	TTCAACAGCTGACGAGCA	
	exon 17,18	TTGGTGAATGCAAGGGAGAA	ACCTTCCGGATGATGTTGT	
	exon 19,20	CTCTATGGCTCTTCTCTACAT	AGTCAACCCACACCTACACCT	
<i>C20orf194</i>	exon 2	TTAAGAAAGTGGGGTCCCTGT	TGAGCCGTTCAGCAAGAA	
	exon 3	ATCCCAAGCAAGTCAATCCT	ACAATTTGGGGGAAACAAG	
	exon 4	GTTCTGGCAAAAAGTGGT	ACTGGGGCTTTGGACTATTT	
	exon 5	TTTGGTTTACAAGGCAAGTGT	TCCTTTTGTCTCCAGGGCATA	
	exon 6	CTTTGGAAACAAGTCCCTGGGT	TTGCAAGAAAAGTCCCAT	
	exon 7	TTTTGCTAGTGGAGCCCT	TAACCTGCTCCATGCCCTGT	
	exon 9	TCTGTATGTGATGTGTGTATGT	TGACCGGAGCAAACTAAATATCC	
	exon 10	GAGATTCATCCATGAGGATGCA	GGGGGAACTACTTTATGGTAT	
	exon 11	CACCTCTCTGAGCATGTGTGTGA	GCAGAGAAACAGACATTTACAG	
	exon 12	CTAGCTTGAAGTTAGTATGTCCC	GGGTTAAATCAACACTAACTGG	
	exon 13	CCATCTCATGACAGAGTAAAGAC	ATCCAGAAATATCAAGAGGGAG	
	exon 14	TGGGGCAGAAAGCTAAACTG	TTATGTTGCCAGGCTGGT	
	exon 15	GCCCAAGCATGTGACTTTT	TGGTGTCAAAGAGCCCAA	
	exon 17	TCATGCCTCAAGTATGACAT	TTTGGCTTCTAAGTGAAGGCTG	
	exon 18,19	AAGGATGACACACCTCACTGT	TGGGATGAGCTGAGAGATCA	
	exon 20	AAAGTCACTCCAGTCAAGAA	AGTGCAGTCTGTGATCATCA	
	exon 21	AGCTCCTGAGAAAGGCAATTT	AACAGCTAGTCAAGGCTGACAT	
	exon 22	ACATGGAATGCTGGAGAGGA	AGGGGAGGAAATGCAAAATGAA	
	exon 23	ATACATTTGGGCAATCTGAGCCT	TCACTCAAAAAGTGGGTGGT	
	exon 24	CACAGGCTTCAAGTCAAAATC	TGTGCTGGTCTGACACTG	
exon 25	AGCACATCTAATCTGAACACAG	ATTAGAAAGCAATGCAACCCCA		
exon 26	TCAAGCCTCTATTTTGAAGCA	GCAAGTGGCAAGATGAA		
exon 27,28	GTGTTTGGAAAGTGTGACTCTG	AAGGAAAGTGGAGTCTGTAA		
exon 29	TGGCTAAAGTCAAGAGTATGT	TACAGGAAGTGTCAAGAGGACT		
exon 30	CCAAGATGGCTTCTCTGAAATG	CTGCTCATGATGATGACTGT		
exon 31	AGATTGGGGCTCTTCTTTT	TCCAAAGGGGTCTATGAGGA		
exon 32	CATGGCTATCCTATGTGCTCAGT	GCTCTTGGAAAGAAATGTCCTA		
exon 33	TATTGGGTGTAAAGGTGTGTCT	TGGCTCAAAAAGTCACTTCTCC		
exon 34	ATTACCTGGGTATGATGGCACA	TTGCTGCTACAGGATGATT		
exon 35,36	GAGGGGTTCACCTTGTGTAG	TTTCTGAGATCTGAGCAGCAT		
exon 37	ACCTACTCCTCCTTCTAAGCTG	TCTCTCAAAGTGTCCCTGCAA		
exon 38	TTATATCCAGGCCATAAGGCA	AATCTGTACGCACTTCCCT		
exon 39	AGGCAAGGCTTGAAGTGT	TTTGGCTGTGCAATCCT		
exon 40	ACAAGAGTCCATCAGGTCCCT	ACAACATGCACTCAAGCCA		

Table S2. Variants Identified by Candidate Gene Sequencing

Gene	Position (NCBI 37.1)		region	position		rs	SNPs	Wild-type	Ped1_IV-4	Ped3_III-1	Ped2_II-1	Ped4_II-1	Ped5_II-1
	Start	End		NT_011387.8	Chr (V37.1)								
<i>PDYN</i> NM_024111.2	1959402	1974702							No variation	No variation	No variation	No variation	No variation
<i>STK5</i> NM_080836.3	2082528	2129201	exon 1	2022732	2082732	rs6112857	Arg69Gly	CC	GG	GC			
				2022767	2082767	rs6106228	Gln80Gln	GG	AG	AA			
				2037688	2097688	rs1891227	Ala423Ala	TT	CT	CC			
<i>TGM3</i> NM_003245.3	2276613	2321725	intron 12	2255929	2315929	rs2076406		GG	AA	AA			
			exon 7	2237790	2297790	rs214814	Ser249Asn	GG	GG	GA			
<i>TGM6</i> NM_198994.2	2361554	2413399	5' near gene	2301505	2361505	rs9680025	5' near gene	AA	AG	GG	AG	AG	GG
			intron 1	2301684	2361684	rs2422753	IVS1+63	CC	CT	CC	CT	CC	CC
			exon 2	2315262	2375262	rs2076405	M58V	TT	TC	CC	CC	TC	CC
			intron 2	2315440	2375440	rs7266902	IVS2+169	GG	GT	TT	GT	GT	TT
			exon 6	2320323	2380323	rs6114033	Lys263Lys	GG	GG	GA	GA	GG	GG
			intron 6	2320396	2380396	rs2076404	IVS6+12	CC	TT	CT	CT	CT	CT
				2320628	2380628	-	IVS6+242	CC	CC	CC	CC	CC	CC
				2320629	2380629	-	IVS6+243	GG	GA	GG	GG	GG	GG
			intron 8	2324151	2384151	rs6137891	IVS8+5	GG	GA	GA	GA	GA	GA
				2338017	2398017	rs2295077	Lys492Lys	CC	CT	CT	CT	TT	CT
			intron 2	2351368	2411368	rs2076648	IVS12+121	CC	CC	CC	CC	CC	CC
				2351737	2411737	rs11470465	IVS12+64	GA/GA	-GA	-GA	-GA	-GA	GA/GA
				2353125	2413125	rs2076653	IVS12-11	GG	GA	GA	GA	GA	GG
				2353126	2413126	rs6036467	IVS12-10	CC	TT	TT	TT	TT	TT
				2353320	2413320	rs2076652	3'-UTR	AA	AG	AG	AG	AG	GG
exon 13	2353472	2413472	rs45610835	3'-UTR	TT	TT	TT	TT	TT	TG			
<i>SNRPB</i> NM_003091.3	2442281	2451499	5' near gene	2391504	2451504	rs6049290	5' near gene	CC	CC	TC			
			5' near gene	2391503	2451503	rs4815262	5' near gene	AA	AA	GA			
			exon 1	2391451	2451451	rs6049288	5'-UTR	GG	GG	TG			
			intron 3	2384665	2444665	rs73606142	IVS3-120	TT	CT	TT			
<i>SNORD119</i> NR_003684.1	2443598	2443693						No variation	No variation				
<i>ZNF343</i> NM_024325.4	2462463	2489778	intron 3	2414176	2474176	-	IVS3-11	TT	TT	TC			
			intron 4	2413587	2473587	rs41308639	IVS4-22	AA	AT	AT			
			exon 7	2403921	2463912	-	Leu65Leu	TT	TT	TA			
<i>ZMC2</i> NM_080751.2	2517253	2622430	exon 3	2539387	2479387	rs6050063	Arg123Lys	AA	AG	AG			
			intron 3	2539568	2479568	rs4815320	IVS3+148	TT	TA	TA			
			intron 4	2542669	2482669	rs7270277	IVS4+13	CC	CT	CC			
				2542747	2482747	rs4815323	IVS4+91	GG	GT	GG			
			intron 6	2552926	2492926	rs1883980	IVS6+11	AA	AG	GG			
				2559778	2499778	rs6083735	IVS6-14	GG	GC	CC			
			intron 9	2572816	2512816	rs6050433	IVS9-139	TT	TG	GG			
			intron 13	2591005	2531009	rs1883978	IVS13-59	TT	GG	GG			
			exon 14	2591232	2531232	rs6050576	Asp527Asp	CC	TT	TT			
			intron 15	2593006	2533006	rs1015159	IVS15+20	AA	AG	GG			
			exon 16	2593863	2533869	rs6515646	Ser589Ser	TT	TC	TC			
				2594254	2534254	rs6115181	non-coding	AA	AG	AG			
			intron 16	2596762	2536762	rs6050622	IVS16-21	GG	GA	GA			
			intron 17	2596969	2536969	rs4621228	IVS17+118	TT	TT	TA			
			exon 18	2597978	2537978	rs4815428	non-coding	GG	GG	GA			
				2598019	2538019	rs6083566	non-coding	CC	CC	CT			
				2598405	2538405	rs13040075	non-coding	GG	GA	GA			
			intron 20	2616556	2556556	rs910271	IVS20-26	TT	TC	CC			
			intron 21	2616679	2556679	rs2422808	IVS21+29	CC	CG	GG			
				2616776	2556776	rs13038659	IVS21+126	TT	TG	TG			
			exon 22	2618094	2558094	rs6037181	IVS21-26	GG	GG	AA			
				2618140	2558140	rs6083915	Ser802Ser	TT	TT	CC			
			intron 22	2618308	2558308	rs6050771	IVS22+71	CC	CC	TT			
exon 23	2561998	2621998	rs6050798	3'-UTR	TT	TT	CT						
<i>NOP56</i> NM_006392.2	2633254	2639039	exon 1	2573296	2633296	rs6138678	5'-UTR	GG	GG	CG	GG	GG	GG
			intron 1	2573397	2633397	rs68063608	IVS1-25	g.263397_263403[S]	g.263397_263403[G]+(2300)	g.263397_263403[R]+(2200)	g.263397_263403[G]+(2200)	g.263397_263403[G]+(2000)	g.263397_263403[R]+(1700)
			exon 9	2577071	2637071	rs8958	Thr345Thr	TT	CT	TT	CT	CT	TT
<i>MIR1292</i> NR_031699.1	2633423	2633488						No variation	No variation	No variation	No variation	No variation	
<i>SNORD110</i> NR_003078.1	2634858	2634932						No variation	No variation				
<i>SNORA51</i> NR_002981.1	2635713	2635844						No variation	No variation				
<i>SNORD86</i> NR_004399.1	2636743	2636828						No variation	No variation				
<i>SNORD56</i> NR_002739.1	2637270	2637340						No variation	No variation				
<i>SNORD57</i> NR_002738.1	2637585	2637656						No variation	No variation				
<i>IDH3B</i> NM_174856.1	2639041	2644843	intron 2	2584407	2644407	rs2073193	IVS2-3	GG	CC	CC			
<i>EBF4</i> NM001110514.1	2673524	2740754	intron 1	2614174	2674174	rs55820831	IVS1-133	AA	AG	AG			
			intron 2	2617329	2677329	rs874688	IVS2-11	TT	TC	CC			
			exon 3	2617566	2677566	rs2325900	non-coding	TT	TT	TC			
			intron 3	2618202;2618203	2678202;2678203	rs11474226	IVS3+58	-/-	-AAAG	AAAGAAAG			
			intron 12	2670870	2730870	rs6138883	IVS12+236	CC	GC	CC			
			intron 15	2672934	2732934	rs60014511	IVS15+49	GG	AG	GG			
			exon 17	2676104	2736104	rs13042767	non-coding	GG	CG	GG			
<i>CPXM1</i> NM_019609.4	2774715	2781292	intron 6	2717828	2777828	rs742707	IVS6+10	GG	AG	GG			
<i>C20orf141</i> NM_080739.2	2795657	2735657	exon 1	2736007	2796007	rs12625619	Leu59Leu	GG	AG	AA			
<i>FAM113A</i> NM_022760.3	2815971	2821332	exon 4	2758801	2818801	rs751899	non-coding	TT	TC	CC			
				2758480	2818480	rs2325970	non-coding	AA	AG	GG			
				2757100	2817100	rs78139021	non-coding	TT	TG	TT			
				2756821	2816821	rs2274669	Pro372Pro	CC	CT	CT			

VPSI6 NM_022575.2	2821373	2847378	exon 3	2780773	2840773	rs3818605	Ser72Ser	CC	CT	CT
			intron 10	2782685	2842685	rs6051449	IVS10-15	AA	AC	CC
			intron 11	2783053	2843053	rs632080	IVS11-49	AA	AG	GG
			intron 16	2784983	2844983	-	IVS16-28	AA	AC	AC
			intron 17	2785130	2845130	rs730819	IVS17-22	GG	GA	AA
PTPRA NM_002836.3	2844841	3019315	intron 3	2785130	2845130	rs730819	IVS3+22	GG	AG	AA
			intron 26	2936423	2996423	rs1178015	IVS26-73	TT	AA	AA
			exon 27	2936497	2996497	rs1178016	Gly303Gly	CC	TT	TT
			intron 27	2936589	2996589	rs1178017	IVS27-58	GG	TT	TT
			intron 31	2942889	3002889	rs544090	IVS31+23	TT	GG	GG
				2943027	3003027	rs2277756	IVS31+161	TT	CT	CT
			intron 37	2957674	3017674	rs561843	IVS31-126	TT	GG	GG
GNRH2 NM_001501.1	3024268	3026391	exon 2	2965107	3025107	rs6051545	Ala16Val	CC	CT	CT
			intron 3	2966238	3026238	rs6138994	IVS3-94	GG	GT	GT
				2966359-	3026359-	rs71195814	frameshift	-/-	-CCCCG	-CCCCG
			exon 4	2966415	3026415	rs8184100	3'-UTR	CC	CT	CT
MRPS26 NM_030811.3	3026675	3028896	intron 2	2967220	3027220	rs2277757	IVS3-40	GG	GA	GA
OLT NM_000915.2	3052266	3053162						No variation	No variation	
AVP NM_000490.4	3063202	3065370						No variation	No variation	
UBOX5 NM_014948.2	3088219	3140540	exon 7	3030848	3090848	rs708973	Arg510Arg	TT	TT	TG
FASTKD5 NM_021826.4	3127165	3140532	exon 2	3068403	3128403	rs3746698	Gly438Gly	AA	AG	AA
ProSAP1P1 NM_014731.2	3143273	3149207	exon 1	3087468	3147468	rs17853865	Asn114Asn	GG	GG	GA
DDRAG1 NM_023935.1	3171012	3185295	intron 1	3087024	3147024	rs7260750	IVS1-13	AA	AA	AG
			intron 4	3124134	3184134	rs2295553	IVS1-73	CC	TC	CC
			intron 4	3116072	3176072	rs7263489	IVS4-73	CC	AC	CC
			intron 5	3115556	3175556	rs2295549	IVS5-42	CC	GC	CC
			intron 7	3112246	3172246	rs2295547	IVS7-16	TT	GT	TT
ITPA NM_033453.2	3190056	3204506	5' near gene	3130039	3190039	rs45620433	5' near gene	CC	CG	CC
			exon 3	3133978	3193978	rs8362	Gln46Gln	GG	GA	GG
			intron 5	3144176	3204176	rs75609817	IVS5-17	GG	GA	AA
			exon 8	3144084	3204084	rs9101	Gln87Glu	GG	GA	GG
SLCAA11 NM_001174089.1	3208063	3219887	5' near gene	3159894	3219894	rs6107260	5' near gene	TT	TC	CC
			exon 1	3159698	3219698	-	5'-UTR	GG	GG	GC
			exon 1	3159692	3219692	rs6084314	5'-UTR	GG	GA	AA
			exon 2	3158634	3218634	rs3810562	Pro26Arg	CC	CG	GG
			exon 3	3158355	3218355	rs3827076	non-coding	GG	GC	CC
			intron 3	3155548	3215548	rs3803957	IVS3-8	GG	GT	GT
			intron 5	3155088	3215088	rs6133022	IVS5+126	GG	GA	AA
			exon 6	3154819	3214819	rs3827075	Arg145Arg	AA	AC	CC
			intron 8	3154126	3214126	rs3803955	IVS8+34	GG	GA	GA
			intron 10	3151719	3211719	rs3803953	IVS10-15	AA	AC	AC
			intron 17	3149371	3209371	rs2281575	IVS17-18	CC	CT	CT
			intron 17	3149357	3209357	rs10048856	IVS17-4	GG	GG	GA
C20orf194 NM_001009984.1	3229948	3388255	exon 4	3302033	3362033	rs6051818	Val92Val	AA	AG	AG
			intron 4	3302009	3362009	rs2008730	IVS4+17	AA	AA	AG
			intron 6	3295567	3355567	rs2208030	IVS6+110	AA	AG	AG
			exon 11	3264373	3324373	-	Phe265Leu	CC	CT	CC
			exon 12	3261244	3321244	rs80335486	Thr296Thr	TT	CG	CT
			intron 19	3239054	3299054	rs6139071	IVS19-7	AA	AG	AG
			intron 22	3236279	3296279	rs2236115	IVS22+103	GG	AG	AG
			intron 23	3236428	3296428	-	IVS23-99	-/-	T/-	T/-
			exon 24	3225140	3285140	rs2422864	Arg577Gly	TT	CC	CC
			intron 25	3217602	3277602	rs2236104	IVS25-6	CC	CA	CA
			exon 27	3215239	3275239	rs2281504	Ser85Ser	GG	AG	GG
			intron 34	3180723	3240723	rs6051685	IVS34-49	CC	CG	CG
			intron 37	3176554	3236554	rs117680746	IVS37+98	GG	AG	AG
			exon 38	3175916	3235916	rs2281496	Lys1092Lys	TT	CT	TT
			intron 40	3173142	3233142	rs6037516	IVS40+76	GG	CC	GC

Table S3. Primers Used for Repeat-Primed PCR, Southern Blotting, and RT-PCR

For repeat-primed PCR

Primer name	Primer sequence
Forward primer	TTTCGGCCTGCGTTCGGG
First reverse primer	TACGCATCCCA GTTTGAGACGCA GGCCCA GGCCCA GGCC
Second reverse primer	TACGCATCCCA GTTTGAGACG

For probes for Southern blot analysis.

Primer name	Primer sequence
Forward primer	TTTAAGAGCTTCCAAGGCTGA
Reverse primer	AGTGCCCA CAAGGAAACCGTTA

For quantitation of mouse NOP56 cDNA

Primer name	Primer sequence
mouse NOP56 F	GTTGGCGCTGAAGGAAGTGG
mouse NOP56 R	CTTTGGCACGAGAGTAGCTG

For quantitation of human NOP56 cDNA

Primer name	Primer sequence
human NOP56 cex4F	TTGCCTTGAAAATGCCAAC
human NOP56 cex6R	TGTATTGCGGCCCAATCTT

For investigation of human NOP56 cDNA splicing variants

Primer name	Primer sequence
human NOP56 cex1F	TAGCCGATTGCGAGCCGAA
human NOP56 cex4R	GTTGCCTTGAAAATGCCAA