

Supplementary table 2. Sanger sequencing validation of called variants

Chr.	Position	Strand	Ref.	Variant	Affected gene	T	B	N	To TSS	Forward primer	Reward primer	Reference sequence (hg19)	Resulting sequence	Validation	Allele
chr1	35545021	+	G	GGC	ZMYM1	+	+	+	59	CCAATTACAGGAGCTGGGA	CGAGTAAGCGCGAGGAGTAA	chr1:35544895+35545096	2021CTGCACCGCTGGCG	Yes	Homozygote
chr1	117602913	+	A	G	TTF2	+	+	-	-35	GGGAGGGATTAAAGCTGCAA	TACGCCACCGCAATCAACA	chr1:117602851+117603048	11CTGCATATGGTGGT	Yes	Homozygote
chr1	26644398	+	C	T	CD52	+	+	+	-12	TGTACGACTAGCCCTGCTCC	ATCTTCGTGGCTGTCTTGTT	chr1:26644265+26644499	2351GACTAAATTAGGGC	Yes	Homozygote
chr1	75198762	+	A	C	TYW3	+	+	+	-99,-77,-73	TGCCACACCTTCTCCAACCT	CCTTCATGTTAGCCTCGTCT	chr1:75198702+75198898	1971AGGTAGGCATGTG	Yes	Homozygote
chr1	174968529	+	T	TC	CACYBP	+	+	+	-41	GAAATGGTCGGTGAAAGGGG	TTTTACAGTGTGTGGCGCG	chr1:174968368+174968611	2.CTTATTTTTGTTGCT	Yes	Homozygote
chr1	45965702	+	G	T	CCDC163P;MMA	+	+	+	-153,48	GGGTTGGTTTTGAAAGTCCGA	GGAGACAGCCACACGAAATC	chr1:45965643+45965835	1931ACTCCAATGCCGCA	Yes	Homozygote
chr1	222763248	-	C	T	TAF1A	+	+	+	-52,-13	CCTCGCCCCAATTFCCATTT	TTAGGTATTAGGCAGCGCG	chr1:222763190-222763404	21CCGGAAGCTGCCTTT	Yes	Homozygote
chr1	210501503	+	T	A	HHAT	+	+	+	-92	AGGGCAGAGTTATCCACCTT	GCTTGGCAGAGTTTCCAGA	chr1:210501433+210501591	1CACGACCCTGAGAC	Yes	Homozygote
chr2	39103031	+	G	GC	DHX57;MORN2	+	+	+	-71	CGCAGAAGTGGAAAGACGTAC	CCTCACCCGGGAAGCGAATA	chr2:39102909+39103121	2131CGGGTTCGGAAGGC	Yes	Homozygote
chr3	124449251	+	T	C	UMPS	-	+	+	40	GACCCATCCAGAAAGAACCC	GCCTGTTTGCTTCAAATTC	chr3:124449105+124449307	2 CCGGGCGCTGGCG	Yes	Homozygote
chr3	15469000	-	A	C	EAF1;METTL6	+	+	+	-63	AGGAATCTGAGGAGGCAAGG	GGAGGCAGAATACGGGAAGA	chr3:15468934-15469106	173b GTCACCCTGCATGC	Yes	Homozygote
chr3	156392134	+	G	A	TIPARP	+	+	+	-70	GTGCTGGGTCTTCTCGGAA	AGTTCGTGGCGTGGACTTA	chr3:156392031+156392210	1 GTCTATCTGCACA	Yes	Heterozygote
chr4	8582233	+	C	T	GPR78	+	+	+	18	TCTTCCCAATTCATCCCGC	GTGGCAGGATGGGACTCTG	chr4:8582084+8582320	237bp GTTAACGGCGGAGC	Yes	Homozygote
chr5	140474189	+	T	TC	PCDHB2	+	+	+	-1	CGCTGAAAGAAAGCACTGGG	CGCCTTAGAAAGCCCTCCTCT	chr5:140474074+140474249	1 TTCTAGAGTCGAGA	Yes	Homozygote
chr5	137673166	+	A	C	FAM53C	+	+	+	-57	TCTGTAACTGGTTCCTCCTC	TCTGTAATTTGGCTACACC	chr5:137673084+137673324	2AATGAAACACGAA	Yes	Homozygote
chr6	122793032	-	A	G	PKIB;SERINC1	+	+	+	-29	CAGGAAAGAAAGCAGCCGTG	TCACAAGAGCAGCGGATACA	chr6:122792892-122793107	21 CCAAAATCGCTATG	Yes	Homozygote
chr6	41196039	+	T	C	TREML4	+	+	+	-22	GAAAGTTCAGTAAAGCGCGC	GTTCACAGTGTCTCTCAGCT	chr6:41195875+41196073	1991GTTGAGTAGACTGA	Yes	Homozygote
chr6	41196040	+	C	T	TREML4	+	+	+	-21	CAGACCCCTGTAGCTGGTGG	GTTTCTGACAGCGGAGGAGA	chr6:41195934+41196151	218 CGGGCGCTACAG	Yes	Homozygote
chr6	41196094	+	C	T	TREML4	+	+	+	34	GACCATTTCCCCAGCAAGAC	CCATTCCAGCCAGATCTGT	chr6:41196013+41196169	157GTTAAGTACAGCT	Yes	Homozygote
chr6	41196103	+	C	CCTT	TREML4	+	+	+	43	CAGCCCTGTAGCTGGTGG	GTTTCTGACAGCGGAGGAGA	chr6:41195934+41196151	218 CCTTTTAGGCTGCC	Yes	Homozygote
chr6	41196155	+	C	T	TREML4	+	+	+	95	AGCTGAGAGACACTGGGAA	TGCACCTTTCCCTCCATGT	chr6:41196054+41196251	198 TTACTATGCAGATC	Yes	Homozygote
chr7	140396471	+	C	CG	NDUF82	+	+	+	-9	GCAGCTCAGCAACCATAAAGT	TGCCTACTTCGCTTCCTCC	chr7:140396323+140396502	1 TCCTAGACGTGCTG	Yes	Homozygote
chr8	30601610	+	TC	T	UBXN8	+	+	+	-71	AGGCTCTCTGGACATCATG	CGGGAAGGAAAGTGGAACTCT	chr8:30601467+30601651	185 CCATAGCACTGCTT	Yes	Homozygote
chr9	37753723	+	C	G	RGNMTD3	+	+	+	-76	ACAGGGTTCCAGAGAGGTTG	CGCTTCGCTTCGTAAGCTC	chr9:37753617+37753787	171 TTCAAATCCGGGAG	Yes	Homozygote
chr10	102133332	+	CT	C	NCRNA00263	+	+	+	0	GCTTCTGATTCCTCTGCTGCT	GAGTCTGGAGCACTGTTCG	chr10:102133242+102133457	ACGGAGCCGGTCTT	Yes	Homozygote
chr10	88428302	+	G	A	LDB3	+	+	+	-123,-18,98	CGAGATATCAGTTCGATGGC	CCAGTCAGGGTACACTGTA	chr10:88428233+88428474	24 ATGTTGCATCTTAGC	Yes	Heterozygote
chr11	66278058	+	C	G	BBS1	+	+	+	-60	CTCTGACTGGGAAGGAGCAA	CGTAACGCCAATAGTGCC	chr11:66277928+66278092	16 AAGATTGAGATCTG	Yes	Homozygote
chr11	118889246	+	A	AG	MIR3656;RPS25	+	+	+	7	TGAGTACTTCCGGGGCAAAA	AAAGTTAGCTGCCGAGACCT	chr11:118889096+118889314	GGACGAGCTGCAC	Yes	Homozygote
chr11	6947627	+	G	CGGGG	ZNF215	+	+	+	-8	GGGGAACAGAAAGGCATTT	CGGATCGGTGAGGTAGACC	chr11:6947488+6947702	215bp AGCACTAGCGAGG	Yes	Homozygote
chr11	93474677	-	G	C	C11orf54;TAF1D	+	+	+	-82	CTCTCAGAAATACCGAGCT	CCTCCAAGTCCCGATAACCA	chr11:93474610-93474835	22b AGGTCCTTAGTCA	Yes	Homozygote
chr11	112831964	+	T	TC	NCAM1	+	+	+	-4	GCTGATTAAGGAAAGCTGGG	TGCCGAACATCAAGGAGGTA	chr11:112831812+112831997	GTTACTATCGTCGCA	Yes	Homozygote
chr12	104324147	+	G	A	GNN;HSP90B1	+	+	+	-55,37	CCCCACGAACTCCTCATT	TGAGTCTCAAGTCCCCTTCG	chr12:104324066+104324280	AGGTCGCTTCTGC	Yes	Homozygote
chr12	54393769	+	A	G	HOXC9	+	+	+	-107,24	TGCCTTGGGTCAGAGTTC	CCTTTTCCCACAGCTCAAT	chr12:54393690+54393875	18 CTACGACGCGTCC	Yes	Homozygote
chr12	54393782	+	G	C	HOXC9	+	+	+	-94,11	TGCCTTGGGTCAGAGTTC	CCTTTTCCCACAGCTCAAT	chr12:54393690+54393875	18 CTATCTGACGGTCCG	Yes	Homozygote
chr14	39583480	+	A	G	SIP1	+	+	+	-7	TAACGCTCCCTAAACTGCCA	CATCAACTCTTCCACTGCGG	chr14:39583427+39583584	15ACTGCGTCTATGTG	Yes	Homozygote
chr14	23299008	+	G	GC	MRPL52	+	+	+	-83	ACGGCTTAATCTGGGAGGAC	AAGCAGAAGTCCGCATCTT	chr14:23298883+23299051	16 GTCTAATCGATGCA	Yes	Homozygote
chr15	85174599	+	G	T	SCAND2	+	+	+	-91	AAGACCCCTCTCGCTCATA	ACTACATTTCCAGAGGCCG	chr15:85174402+85174649	24 CGGGAAGAAGACG	Yes	Homozygote
chr17	46970082	+	G	GA	ATP5G1	+	+	+	-65	TCCAGATCTCGTAGGAAAGCC	CAGACTCACCTCCCAGCTTT	chr17:46969978+46970211	23-GGGGCTAGGAGGA	Yes	Homozygote
chr18	59854475	-	AG	A	KIAA1468;PIGN	+	+	+	-48	CCCAGCTTAGAGACAAGGAGT	TATCTGGCGCTGGGAATGG	chr18:59854399-59854618	22C CTTGGCAGGGGGA	Yes	Homozygote
chr19	23945868	-	C	T	RPSAP58	-	+	+	54	TCTTTACCTCTTGTCTCCCG	CTGGTTTCACAGCTGGTCT	chr19:23945785+23945989	20 CGTGGCTTGGTCC	Yes	Homozygote
chr19	53073604	+	A	G	ZNF701	+	+	+	80	CTGGCTTTAGTCTGTGCG	CTGGTTGAAGGAAAGATGGG	chr19:53073508+53073699	19 CTCGGGGCGACTGAC	Yes	Homozygote
chr19	41903674	+	A	G	BCKDHA;EXOSC	+	+	+	-19	CACAGATCTAGCCAGTCCCC	ACCACAGAACGGGAAGA	chr19:41903608-41903843	23b GTTGGTGGAGATC	Yes	Homozygote
chr21	34697263	+	T	C	IFNAR1	+	+	+	51	GGTGCTAGCTAGGAGGAAAGG	TCCCACAGTTACATGTTTC	chr21:34697140+34697343	20-GATTTACGACGCG	Yes	Homozygote
chr22	42896553	+	TGGGGT	G	SERHL	+	+	+	-27	AGAGATGGCGGTGGAAAGG	CCTGGTGAAGTCTAAGGCGAC	chr22:42896492+42896718	22 CCATTACTGCTGAC	Yes	Homozygote
chrX	119005704	-	A	G	NDUFA1;RNF11	+	+	+	-29	CATCTCTGCCCGGTACTTA	GCAAAAGACACTGACGGAA	chrX:119005671-119005877	2 AAACGTCTGGGTCT	Yes	Homozygote
chr1	192544794	+	C	G	RGS1	+	+	+	-62	CAACCAACAGACCCTGT	ATGCGCTAGTCAACGCTCTC	chr1:192544731+192544906	1 TGTTATACGATACCC	No	
chr4	144106030	+	T	TC	USP38	+	+	+	-39	GGAGAGAGGGAACAAGGGAG	GAGAAGAGACCAAGGCACA	chr4:144105953+144106112	1 CCTGGTCCGCGG	No	
chr6	41196033	+	T	C	TREML4	+	+	+	-28	ATAAAGCAGCAGACATGGGG	CAGTTCCCAAGTGTCTCTCA	chr6:41195854+41196076	223 CGGCTTGCTTTGTA	No	
chr6	134274223	+	T	G	TBPL1	+	+	+	-77	ACCTCTTACCTCCCACCGA	ATAGACTAGACGCTGGGGG	chr6:134274122+134274342	1 CCGTCCCCATCTA	No	
chr15	85174630	+	G	A	SCAND2	+	+	+	-60	CTAAGCTGTGCTCTGCTTTC	CGTGACATGTTCTTACGCGG	chr15:85174603+85174772	17 TCCAAAACATTGGG	No	
chr6	26383385	+	T	C	BTN2A2	+	+	+	-53,58,63	ATCCTGGGCTCTAACTGCT	CCAGACAACAAGCATCCCA	chr6:26383303+26383454	152 PCR failed	PCR failed	
chr12	55413629	+	G	A	NEUROD4	+	+	+	-99	TTACGACACTTCTCCCCT	AGGAGTGATGTTGCCAGAG	chr12:55413544+55413704	16 PCR failed	PCR failed	