S9 Table. Gene signatures of candidate genes

Chromosome	POS	ID	REF seq	ALT seq	Varyty pe	Gene name	Type of variant	cDNA change	AA change	COSMID
chr3	29529913	rs3043392;rs5847572	TTC	T	DEL	RBMS3	Splice region & intron	c.249-7_249-6delCT		
chr17	59482168	rs35619711;rs201348033	GC	G	DEL	TBX2	Intron	c.1051+39delC		
chr6	158993341	rs34964890	CATA	C	DEL	TMEM181	Intron	c.420-1110_420-1108delATA		
chr19	11459665	rs78738753	G	GA	INS	CCDC159	Intron	c.22-664dupA		
chr11	77629132	rs112620226;rs397702235;rs60400274	A	AT	INS	INTS4	Intron	c.1922+734dupA		
chr17	7412965	rs112391124;rs71541942;rs3833903	T	TAGA	INS	POLR2A	Intron	c.3813+17_3813+19dupAAG		
chr16	89590168	rs113476208;rs77381048	T	TATCTC	INS	SPG7	Intron	c.377-245_377-244insTCTCA		
chr19	11459665	rs78738753	G	GA	INS	TMEM205	Upstream gene	c.-3371dupT		
chr11	100859439	rs524811;rs111177640	G	A	SNP	ARHGAP42	Splice region & intron	c.2537-5G>A		
chr19	47763573	rs45591932	C	T	SNP	CCDC9	Splice region & intron	c.109-5C>T		
chr19	47768128	rs2032811	G	C	SNP	CCDC9	Missense	c.645G>C	p.Glu215Asp	COSM3749524
chr19	47774447	rs66726054	G	A	SNP	CCDC9	Splice region & intron	c.1191+3G>A		
chr19	47774772	rs888836	T	C	SNP	CCDC9	Missense	c.1433T>C	p.Leu478Pro	COSM3721172
chr16	71319646	rs3096380	G	A	SNP	CMTR2	Missense	c.178C>T	p.Leu60Phe	
chr17	13980350	rs8077302;rs2072279	G	A	SNP	COX10	Missense	c.476G>A	p.Arg159Gln	
chr16	89655221	rs3794630	G	A	SNP	CPNE7	Splice region & intron	c.1286+5G>A,c.1061+5G>A		
chrX	125299467	rs10126452	G	T	SNP	DCAF12L2	Missense	c.441C>A	p.His147Gln	COSM3759350
chr6	159655383	rs2501176	A	G	SNP	FNDC1	Missense	c.3839A>G	p.Gln1280Arg	COSM4003774
chr6	159654551	rs370434	C	G	SNP	FNDC1	Missense	c.3007C>G	p.Gln1003Glu	COSM3761627
chr6	159655084	rs420054	C	G	SNP	FNDC1	Missense	c.3540C>G	p.Asp1180Glu	COSM4160339
chr6	159652931	rs420137	G	C	SNP	FNDC1	Missense	c.1387G>C	p.Glu463Gln	COSM450819
chr6	159655326	rs3003174	T	C	SNP	FNDC1	Missense	c.3782T>C	p.Leu1261Pro	COSM3749959
chr3	69230061	rs9831516	G	A	SNP	FRMD4B	Missense	c.2840C>T	p.Ser947Leu	

chr6	29910767	rs1136689	G	C	SNP	HLA-A	Missense	c.307G>C	p.Gly103Arg	COSM4160510
chr1	159024668	rs6940	A	T	SNP	IFI16	Missense	c.2167A>T	p.Thr723Ser	
chr19	47778412	rs1055218	G	T	SNP	INAFM1	Missense	c.236G>T	p.Arg79Leu	COSM3756983
chr14	24653523	rs11550452	G	C	SNP	IPO4	Missense	c.1738C>G	p.Pro580Ala	COSM3753915
chr14	24653954	rs7146310	G	A	SNP	IPO4	Missense	c.1538C>T	p.Ala513Val	COSM3753916
chr6	150716533	rs4407723	T	A	SNP	IYD	Missense, intron, intron	c.691T>A,c.688-58T>A,c.687+1142T>A	p.Phe231Ile	COSM150193
chr1	46746164	rs11542623;rs386518841	C	T	SNP	LRRC41	Missense	c.1825G>A	p.Val609Ile	COSM3997555
chr17	73609139	rs3803728	C	T	SNP	MYO15B	Missense	c.4702C>T	p.Arg1568Trp	
chr14	79434693	rs2270964	C	A	SNP	NRXN3	Splice region& intron	c.2023+4C>A,n.4114+4C>A		COSM3999385
chr1	46827456	rs9865	A	G	SNP	NSUN4	Missense	c.1093A>G,c.946A>G,c.946A>G	p.Ile365Val,p.Ile316Val, p.Ile316Val	COSM3997560
chr21	47851753	rs2073376	A	G	SNP	PCNT	Missense	c.8375A>G,c.8021A>G	p.Gln2792Arg,p.Gln2674Arg	COSM4135105
chr21	47851796	rs9983522	G	A	SNP	PCNT	Synonymous_variant, synonymous_variant	c.8418G>A,c.8064G>A	p.Ala2806Ala,p.Ala2688Ala	.
chr21	47850484	rs2070426	G	C	SNP	PCNT	Missense	c.7977G>C,c.7623G>C	p.Gln2659His,p.Gln2541His	COSM3759003
chr11	9087533	rs2003906	A	G	SNP	SCUBE2	Splice region & intron	c.761-6T>C		
chr17	59544863	rs758596;rs386611686	G	A	SNP	TBX4	Splice region & intron	c.402-8G>A		COSM4000245
chr14	23994517	rs4982766	A	G	SNP	ZFHX2	Missense	c.4634T>C	p.Val1545Ala	
chr14	23992742	rs223124	T	G	SNP	ZFHX2	Missense	c.6409A>C	p.Ser2137Arg	

POS, position; DEL, deletion; INS, insertion; SNP, single nucleotide polymorphism; REF, reference allele; ALT, alternative allele.