

Chromosome	Start	End	Zygoty	Reference	SNP Call	SNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr12	102541977	102541977	Heterozygous	T	-	Novel	C12orf48	INTRONIC	
chr12	104138970	104138970	Heterozygous	C	-	Novel	STAB2	SYNONYMOUS_CODING	
chr12	104373728	104373729	Heterozygous	-	A	Novel	TDG	FRAMESHIFT_CODING	
chr12	104714848	104714848	Heterozygous	T	-	Novel	TXNRD1	INTRONIC	
chr12	104714848	104714848	Heterozygous	T	-	Novel	TXNRD1	INTRONIC	
chr12	104714848	104714848	Heterozygous	T	-	Novel	TXNRD1	INTRONIC	
chr12	104714848	104714848	Heterozygous	T	-	Novel	TXNRD1	INTRONIC	
chr12	104714848	104714848	Heterozygous	T	-	Novel	TXNRD1	INTRONIC	
chr12	105440459	105440459	Heterozygous	A	G	Novel		GENIC	
chr12	105538271	105538271	Heterozygous	T	-	Novel	KIAA1033	INTRONIC	
chr12	108138177	108138177	Heterozygous	A	T	Novel	PRDM4	INTRONIC	
chr12	108138184	108138184	Heterozygous	G	A	Novel	PRDM4	INTRONIC	
chr12	10923543	10923543	Heterozygous	G	A	Novel	ACACB	NON_SYNONYMI_E>K	
chr12	109948227	109948227	Heterozygous	G	A	Novel	UBE3B	NON_SYNONYMI_R>Q	
chr12	109948227	109948227	Heterozygous	G	A	Novel	UBE3B	NON_SYNONYMI_R>Q	
chr12	110874504	110874504	Heterozygous	T	A	Novel	ARPC3	INTRONIC	
chr12	111885352	111885356	Heterozygous	TGGGG	-----	Novel	SH2B3	SPLICE_SITE	
chr12	112130610	112130610	Heterozygous	C	T	Novel	ACAD10	STOP_GAINED_R->Stop	
chr12	112130610	112130610	Heterozygous	C	T	Novel	ACAD10	STOP_GAINED_R->Stop	
chr12	112147524	112147524	Heterozygous	A	C	Novel	ACAD10	INTRONIC	
chr12	112147524	112147524	Heterozygous	A	C	Novel	ACAD10	INTRONIC	
chr12	112220903	112220903	Heterozygous	G	A	Novel	ALDH2	INTRONIC	
chr12	112451180	112451181	Heterozygous	-	C	Novel	TMEM118	UPSTREAM	
chr12	112451180	112451181	Heterozygous	-	C	Novel	TMEM118	UPSTREAM	
chr12	112451180	112451181	Heterozygous	-	C	Novel	TMEM118	UPSTREAM	
chr12	112451180	112451181	Heterozygous	-	C	Novel	TMEM118	UPSTREAM	
chr12	112451190	112451191	Heterozygous	-	C	Novel	ERP29	SPRIME_UTR	
chr12	112451190	112451191	Heterozygous	-	C	Novel	ERP29	SPRIME_UTR	
chr12	112481733	112481733	Heterozygous	T	-	Novel	NAA25	INTRONIC	
chr12	112807491	112807491	Heterozygous	A	C	Novel		GENIC	
chr12	112885781	112885782	Heterozygous	-	AC	Novel		GENIC	
chr12	112888167	112888167	Heterozygous	A	C	Novel		GENIC	
chr12	113440985	113440985	Heterozygous	G	A	Novel	OAS2	INTRONIC	
chr12	113440985	113440985	Heterozygous	G	A	Novel	OAS2	INTRONIC	
chr12	114358581	114358581	Heterozygous	G	C	Novel	RBM19	INTRONIC	
chr12	114358581	114358581	Heterozygous	G	C	Novel	RBM19	INTRONIC	
chr12	114358581	114358581	Heterozygous	G	C	Novel	RBM19	INTRONIC	
chr12	117014175	117014175	Heterozygous	A	-	Novel	MAP1LC3B2	SPRIME_UTR	
chr12	117188082	117188082	Heterozygous	C	T	Novel	RNF27	SYNONYMOUS_CODING	
chr12	117188082	117188082	Heterozygous	C	T	Novel	RNF27	SYNONYMOUS_CODING	
chr12	117611893	117611893	Heterozygous	G	A	Novel	FBXO21	INTRONIC	
chr12	117611893	117611893	Heterozygous	G	A	Novel	FBXO21	INTRONIC	
chr12	118473176	118473176	Heterozygous	C	T	Novel	WSB2	INTRONIC	
chr12	118520157	118520157	Heterozygous	A	T	Novel	VSIQ10	NON_SYNONYMI_S>T	
chr12	118615207	118615207	Heterozygous	T	A	Novel	TAOK3	INTRONIC	
chr12	118673483	118673484	Heterozygous	-	A	Novel	TAOK3	SPLICE_SITE	
chr12	118821801	118821801	Heterozygous	T	-	Novel	SUDS3	INTRONIC	
chr12	120150208	120150208	Heterozygous	G	C	Novel	CIT	INTRONIC	
chr12	120582090	120582090	Heterozygous	C	T	Novel	GCN1L1	INTRONIC	
chr12	120612930	120612931	Heterozygous	AA	-	Novel	GCN1L1	INTRONIC	
chr12	121176324	121176324	Heterozygous	C	T	Novel	ACADS	INTRONIC	
chr12	121201678	121201678	Homozygous	A	-	Novel	SPPL3	SPRIME_UTR	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	121701550	121701550	Heterozygous	T	G	Novel	GAMKK2	INTRONIC	
chr12	122017849	122017849	Heterozygous	A	-	Novel	KDM2B	INTRONIC	
chr12	122017849	122017849	Heterozygous	A	-	Novel	KDM2B	INTRONIC	
chr12	122757370	122757371	Heterozygous	TG	---	Novel	CLIP1	SPRIME_UTR	
chr12	122757370	122757371	Heterozygous	TG	---	Novel	CLIP1	SPRIME_UTR	
chr12	122812704	122812704	Heterozygous	C	T	Novel	CLIP1	SYNONYMOUS_CODING	
chr12	122812704	122812704	Heterozygous	C	T	Novel	CLIP1	SYNONYMOUS_CODING	
chr12	123103008	123103009	Heterozygous	-	T	Novel	KNTC1	INTRONIC	
chr12	123253695	123253696	Heterozygous	-	AA	Novel	DENR	SPRIME_UTR	
chr12	123490156	123490156	Heterozygous	G	A	Novel	PITPNM2	INTRONIC	
chr12	124216625	124216625	Heterozygous	C	T	Novel	ATP6V0A2	INTRONIC	
chr12	124317929	124317929	Heterozygous	C	T	Novel	DNAH10	INTRONIC	
chr12	124362256	124362257	Heterozygous	-	G	Novel	DNAH10	INTRONIC	
chr12	129298759	129298759	Heterozygous	C	T	Novel	SLC15A4	INTRONIC	
chr12	129298775	129298775	Heterozygous	C	G	Novel	SLC15A4	INTRONIC	
chr12	129298777	129298777	Heterozygous	G	A	Novel	SLC15A4	INTRONIC	
chr12	129298779	129298780	Heterozygous	CG	TG	Novel	SLC15A4	INTRONIC	
chr12	129298779	129298780	Heterozygous	CG	CA	Novel	SLC15A4	INTRONIC	
chr12	129298797	129298797	Heterozygous	G	A	Novel	SLC15A4	INTRONIC	
chr12	129429823	129429823	Heterozygous	G	A	Novel	GLT1D1	INTRONIC	

Chromosome	Start	End	Zygoty	Reference	SNP Call	SNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr12	130883555	130883555	Heterozygous	T	-	Novel	RIMBP2	3PRIME_UTR	
chr12	131476668	131476668	Heterozygous	A	-	Novel	GPR133	INTRONIC	
chr12	131569000	131569000	Heterozygous	T	C	Novel	GPR133	INTRONIC	
chr12	133258858	133258858	Heterozygous	C	T	Novel	GOLGA3	INTRONIC	
chr12	133258858	133258858	Heterozygous	C	T	Novel	GOLGA3	INTRONIC	
chr12	133363219	133363219	Heterozygous	C	A	Novel	GOLGA3	DOWNS_TREAM	
chr12	133363219	133363219	Heterozygous	C	A	Novel	GOLGA3	INTRONIC	
chr12	133363222	133363222	Heterozygous	C	T	Novel	GOLGA3	INTRONIC	
chr12	133363222	133363222	Heterozygous	C	T	Novel	GOLGA3	INTRONIC	
chr12	133363222	133363222	Heterozygous	C	T	Novel	GOLGA3	INTRONIC	
chr12	133363232	133363232	Heterozygous	A	G	Novel	GOLGA3	INTRONIC	
chr12	133363232	133363232	Heterozygous	A	G	Novel	GOLGA3	INTRONIC	
chr13	19999997	19999997	Heterozygous	T	C	Novel	TPTE2	INTRONIC	
chr13	19999997	19999997	Heterozygous	T	C	Novel	TPTE2	INTRONIC	
chr13	19999997	19999997	Heterozygous	T	C	Novel	TPTE2	INTRONIC	
chr13	20000007	20000007	Heterozygous	G	A	Novel	TPTE2	INTRONIC	
chr13	20000007	20000007	Heterozygous	G	A	Novel	TPTE2	INTRONIC	
chr13	20000007	20000007	Heterozygous	G	A	Novel	TPTE2	INTRONIC	
chr13	20608357	20608357	Heterozygous	C	T	Novel	ZMYM2	INTRONIC	
chr13	20608357	20608357	Heterozygous	C	T	Novel	ZMYM2	INTRONIC	
chr13	20608357	20608357	Heterozygous	C	T	Novel	ZMYM2	INTRONIC	
chr13	20608357	20608357	Heterozygous	C	T	Novel	ZMYM2	INTRONIC	
chr13	25017678	25017679	Heterozygous	-	CA	Novel	PARP4	INTRONIC	
chr13	25416088	25416089	Heterozygous	-	GT	Novel	RNF17	INTRONIC	
chr13	25416088	25416089	Heterozygous	-	GT	Novel	RNF17	INTRONIC	
chr13	25457398	25457398	Heterozygous	C	T	Novel	CENPJ	NON_SYNONYMI_G>S	
chr13	25457538	25457538	Heterozygous	A	-	Novel	CENPJ	NON_SYNONYMI_G>S	
chr13	25534004	25534004	Heterozygous	A	T	Novel	TPTE2P1	INTRONIC	
chr13	25538050	25538050	Heterozygous	T	-	Novel	TPTE2P1	INTRONIC	
chr13	25538053	25538053	Heterozygous	T	G	Novel	TPTE2P1	INTRONIC	
chr13	25839870	25839870	Heterozygous	A	-	Novel	MTMR6	INTRONIC	
chr13	25894827	25894827	Heterozygous	A	T	Novel	NUPL1	INTRONIC	
chr13	25894827	25894827	Heterozygous	A	T	Novel	NUPL1	INTRONIC	
chr13	25898091	25898092	Heterozygous	-	T	Novel	NUPL1	INTRONIC	
chr13	25898091	25898092	Heterozygous	-	T	Novel	NUPL1	INTRONIC	
chr13	25911051	25911051	Heterozygous	T	-	Novel	NUPL1	INTRONIC	
chr13	25911051	25911051	Heterozygous	T	-	Novel	NUPL1	INTRONIC	
chr13	25911051	25911051	Heterozygous	T	-	Novel	NUPL1	INTRONIC	
chr13	25911051	25911051	Heterozygous	T	-	Novel	NUPL1	INTRONIC	
chr13	26445406	26445406	Heterozygous	T	A	Novel	ATP8A2	NON_SYNONYMI_L>F	
chr13	28133957	28133957	Heterozygous	A	-	Novel	LNR2	INTRONIC	
chr13	28600313	28600313	Heterozygous	G	A	Novel	MTUS2	NON_SYNONYMI_R>H	
chr13	32848908	32848908	Heterozygous	C	-	Novel	FRY	INTRONIC	
chr13	33012742	33012742	Heterozygous	A	G	Novel	NMBP2L2	INTRONIC	
chr13	36700339	36700340	Heterozygous	TA	-	Novel	DCLK1	INTRONIC	
chr13	39542743	39542743	Heterozygous	T	C	Novel	STOML3	INTRONIC	
chr13	39542743	39542743	Heterozygous	T	C	Novel	STOML3	INTRONIC	
chr13	39542743	39542743	Heterozygous	T	C	Novel	STOML3	INTRONIC	
chr13	39550715	39550715	Heterozygous	C	T	Novel	STOML3	NON_SYNONYMI_R>H	
chr13	39550715	39550715	Heterozygous	C	T	Novel	STOML3	NON_SYNONYMI_R>H	
chr13	41034385	41034386	Heterozygous	-	A	Novel	LOC646982	INTRONIC	
chr13	41034385	41034386	Heterozygous	-	A	Novel	LOC646982	INTRONIC	
chr13	41034385	41034386	Heterozygous	-	A	Novel	LOC646982	INTRONIC	
chr13	41330890	41330890	Heterozygous	T	C	Novel	MRPS31	INTRONIC	
chr13	42734107	42734107	Heterozygous	G	A	Novel	DGKH	INTRONIC	
chr13	42734107	42734107	Heterozygous	G	A	Novel	DGKH	INTRONIC	
chr13	42780317	42780317	Heterozygous	A	C	Novel	DGKH	INTRONIC	
chr13	42780317	42780317	Heterozygous	A	C	Novel	DGKH	INTRONIC	
chr13	42788584	42788584	Heterozygous	A	G	Novel	DGKH	INTRONIC	
chr13	42788584	42788584	Heterozygous	A	G	Novel	DGKH	INTRONIC	
chr13	42785721								

Chromosome	Start	End	Zygoty	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr13	77531083	77531083	Heterozygous	T	C	Novel		GENIC	
chr13	78211390	78211390	Heterozygous	T	C	Novel	SCEL	INTRONIC	
chr13	78211390	78211390	Heterozygous	T	C	Novel	SCEL	INTRONIC	
chr13	78211390	78211390	Heterozygous	T	C	Novel	SCEL	INTRONIC	
chr13	78327545	78327545	Heterozygous	T	A	Novel	SLAIN1	INTRONIC	
chr13	78327545	78327545	Heterozygous	T	A	Novel	SLAIN1	INTRONIC	
chr13	98552957	98552957	Heterozygous	C	A	Novel	UGGT2	INTRONIC	
chr13	99105322	99105322	Heterozygous	C	T	Novel	STK24	3PRIME_UTR	
chr13	99105322	99105322	Heterozygous	C	T	Novel	STK24	3PRIME_UTR	
chr13	99174002	99174002	Heterozygous	T	G	Novel	STK24	INTRONIC	
chr13	99174002	99174002	Heterozygous	T	G	Novel	STK24	INTRONIC	
chr13	101020758	101020758	Heterozygous	G	T	Novel	PCCA	NON_SYNONYMIW->L	
chr13	101020758	101020758	Heterozygous	G	T	Novel	PCCA	NON_SYNONYMIW->L	
chr13	101020758	101020758	Heterozygous	G	T	Novel	PCCA	NON_SYNONYMIW->L	
chr13	101192483	101192483	Heterozygous	T	A	Novel		GENIC	
chr13	102235765	102235765	Heterozygous	A	-	Novel	ITGBL1	INTRONIC	
chr13	108518715	108518715	Heterozygous	C	T	Novel	FAM155A	NON_SYNONYMIW->Q	
chr13	109717875	109717875	Heterozygous	A	-	Novel	MYO18	INTRONIC	
chr13	110864130	110864130	Heterozygous	AAAG	---	Novel	COL4A1	INTRONIC	
chr13	113532640	113532640	Heterozygous	G	A	Novel	ATP11A	INTRONIC	
chr13	113532640	113532640	Heterozygous	G	A	Novel	ATP11A	INTRONIC	
chr13	115002098	115002098	Heterozygous	T	-	Novel	CDC16	INTRONIC	
chr13	115002098	115002098	Heterozygous	T	-	Novel	CDC16	INTRONIC	
chr13	115051988	115051988	Heterozygous	G	A	Novel	UPF3A	SPLICE_SITE	
chr13	115051988	115051988	Heterozygous	G	A	Novel	UPF3A	SPLICE_SITE	
chr14	19117185	19117185	Heterozygous	C	A	Novel		GENIC	
chr14	19118232	19118232	Heterozygous	C	G	Novel		GENIC	
chr14	19118254	19118254	Heterozygous	T	A	Novel		GENIC	
chr14	19118308	19118308	Heterozygous	T	C	Novel		GENIC	
chr14	19553494	19553494	Heterozygous	G	A	Novel	POTEG	SYNONYMOUS_CODING	
chr14	19553494	19553494	Heterozygous	G	A	Novel	POTEG	EXONIC	
chr14	19808550	19808550	Heterozygous	A	T	Novel		GENIC	
chr14	20146338	20146338	Heterozygous	C	T	Novel		GENIC	
chr14	20216010	20216010	Heterozygous	T	C	Novel	OR4Q3	EXONIC	
chr14	20528309	20528309	Heterozygous	G	A	Novel	OR4L1	NON_SYNONYMIW->S	
chr14	20873540	20873540	Heterozygous	G	A	Novel	TEP1	INTRONIC	
chr14	20917333	20917333	Heterozygous	C	T	Novel	OSGPE	SYNONYMOUS_CODING	
chr14	21500105	21500105	Heterozygous	G	A	Novel	RNASE13	DOWNSTREAM	
chr14	21931333	21931333	Heterozygous	---	GGT	Novel	SALL2	NON_SYNONYMOUS_CODING	
chr14	2294951	2294951	Heterozygous	A	G	Novel		GENIC	
chr14	23013105	23013105	Heterozygous	G	A	Novel		GENIC	
chr14	23371570	23371570	Heterozygous	G	A	Novel	RBM23	NON_SYNONYMIW->W	
chr14	23371570	23371570	Heterozygous	G	A	Novel	RBM23	NON_SYNONYMIW->W	
chr14	23371570	23371570	Heterozygous	G	A	Novel	RBM23	NON_SYNONYMIW->W	
chr14	23524790	23524790	Heterozygous	G	A	Novel	CDH24	SYNONYMOUS_CODING	
chr14	23524790	23524790	Heterozygous	G	A	Novel	CDH24	SYNONYMOUS_CODING	
chr14	23548776	23548777	Heterozygous	CT	GA	Novel	ACIN1	NON_SYNONYISA->SP	
chr14	23548776	23548777	Heterozygous	CT	GA	Novel	ACIN1	NON_SYNONYISA->SP	
chr14	23548776	23548777	Heterozygous	CT	GA	Novel	ACIN1	NON_SYNONYISA->SP	
chr14	23612514	23612514	Heterozygous	T	C	Novel	SLC7A8	INTRONIC	
chr14	23612514	23612514	Heterozygous	T	C	Novel	SLC7A8	INTRONIC	
chr14	23744801	23744803	Heterozygous	CAT	---	Novel	HOMEZ	NON_SYNONYMOUS_CODING	
chr14	24112441	24112441	Heterozygous	G	A	Novel	DHRS2	INTRONIC	
chr14	24112441	24112441	Heterozygous	G	A	Novel	DHRS2	INTRONIC	
chr14	24424224	24424224	Heterozygous	T	G	Novel	C14orf167	UPSTREAM	
chr14	24424224	24424224	Heterozygous	T	G	Novel	C14orf167	UPSTREAM	
chr14	24424224	24424224	Heterozygous	T	G	Novel	C14orf167	EXONIC	
chr14	24424224	24424224	Heterozygous	T	G	Novel	C14orf167	UPSTREAM	
chr14	24424224	24424224	Heterozygous	T	G	Novel	DHRS4	INTRONIC	
chr14	24435672	24435672	Heterozygous	A	C	Novel	DHRS4	INTRONIC	
chr14	24505640	24505640	Heterozygous	C	T	Novel	DHRS4L1	UPSTREAM	
chr14	24511866	24511866	Heterozygous	G	A	Novel	DHRS4L1	INTRONIC	
chr14	25076494	25076494	Heterozygous	C	T	Novel	GZMH	NON_SYNONYIS->N	
chr14	29261305	29261305	Heterozygous	A	-	Novel	C14orf23	EXONIC	
chr14	31617919	31617919	Heterozygous	A	G	Novel	HECTD1	SPLICE_SITE	
chr14	31618073	31618073	Heterozygous	C	-	Novel	HECTD1	INTRONIC	
chr14	31625994	31625995	Heterozygous	-	A	Novel	HECTD1	INTRONIC	
chr14	31625995	31625995	Heterozygous	T	C	Novel	HECTD1	INTRONIC	
chr14	32142544	32142545	Heterozygous	-	T	Novel	NUPBL	INTRONIC	
chr14	35242710	35242710	Heterozygous	A	-	Novel	BAZ1A	INTRONIC	
chr14	35242710	35242710	Heterozygous	A	-	Novel	BAZ1A	INTRONIC	
chr14	36121089	36121089	Heterozygous	A	-	Novel	RALLGAP1	INTRONIC	
chr14	36121088	36121088	Heterozygous	A	-	Novel	RALLGAP1	INTRONIC	
chr14	36334908	36334908	Heterozygous	T	-	Novel	BRMS1L	INTRONIC	
chr14	36334908	36334908	Heterozygous	T	-	Novel	BRMS1L	INTRONIC	
chr14	38310733	38310733	Heterozygous	A	G	Novel		GENIC	
chr14	39602850	39602850	Heterozygous	T	-	Novel	SIP1	INTRONIC	
chr14	39602850	39602850	Heterozygous	T	-	Novel	SIP1	INTRONIC	

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chr14	39602850	39602850	Heterozygous	T	-	Novel	SIP1	INTRONIC	
chr14	39796049	39796049	Heterozygous	T	-	Novel	CTAGES	INTRONIC	
chr14	39796049	39796049	Heterozygous	T	-	Novel	CTAGES	INTRONIC	
chr14	39796049	39796049	Heterozygous	T	-	Novel	CTAGES	INTRONIC	
chr14	39796049	39796049	Heterozygous	T	-	Novel	CTAGES	INTRONIC	
chr14	45564466	45564466	Heterozygous	A	T	Novel	PRPF39	NON_SYNONYIE->D	
chr14	50295722	50295722	Heterozygous	A	G	Novel	SDCCAG1	INTRONIC	
chr14	50299052	50299052	Heterozygous	T	-	Novel	SDCCAG1	INTRONIC	
chr14	50605267	50605268	Heterozygous	-	T	Novel	SOS2	INTRONIC	
chr14	50605507	50605508	Heterozygous	-	A	Novel	SOS2	SPLICE_SITE	
chr14	50895476	50895476	Heterozygous	A	-	Novel	MAP4K5	SPLICE_SITE	
chr14	50895476	50895476	Heterozygous	A	-	Novel	MAP4K5	SPLICE_SITE	
chr14	51219249	51219249	Heterozygous	C	T	Novel	NIN	NON_SYNONYIR->H	
chr14	51219249	51219249	Heterozygous	C	T	Novel	NIN	NON_SYNONYIR->H	
chr14	51219249	51219249	Heterozygous	C	T	Novel	NIN	NON_SYNONYIR->H	
chr14	51219501	51219501	Heterozygous	A	-	Novel	NIN	NON_SYNONYIR->H	
chr14	51219501	51219501	Heterozygous	A	-	Novel	NIN	INTRONIC	
chr14	51219501	51219501	Heterozygous	A	-	Novel	NIN	INTRONIC	
chr14	51219501	51219501	Heterozygous	A	-	Novel	NIN	INTRONIC	
chr14	51348385	51348385	Heterozygous	T	C	Novel	ABHD12B	INTRONIC	
chr14	51348385	51348385	Heterozygous	T	C	Novel	ABHD12B	INTRONIC	
chr14	51710543	51710544	Heterozygous	-	T	Novel	TMX1	INTRONIC	
chr14	51716162	51716162	Heterozygous	T	-	Novel	TMX1	INTRONIC	
chr14	52957732	52957732	Heterozygous	A	-	Novel	TXNDC16	INTRONIC	
chr14	52957732	52957732	Heterozygous	A	-	Novel	TXNDC16	INTRONIC	
chr14	55005126	55005126	Heterozygous	A	G	Novel	CGRFR1	3PRIME_UTR	
chr14	55169339	55169339	Homozygous	G	A	Novel	SAMD4A	INTRONIC	
chr14	55169339	55169339	Homozygous	G	A	Novel	SAMD4A	INTRONIC	
chr14	57113896	57113896	Heterozygous	T	A	Novel	C14orf101	INTRONIC	
chr14	57678824	57678824	Homozygous	T	-	Novel	EXOC5	INTRONIC	
chr14	58825826	58825826	Heterozygous	T	-	Novel	ARID4A	INTRONIC	
chr14	58825826	58825826	Heterozygous	T	-	Novel	ARID4A	INTRONIC	
chr14	58825826	58825826	Heterozygous	T	-	Novel	ARID4A	INTRONIC	
chr14	60485027	60485027	Heterozygous	C	-	Novel		GENIC	
chr14	61446808	61446808	Heterozygous	A	-	Novel	TRMT5	SPLICE_SITE	
chr14	61446808	61446808	Heterozygous	A	-	Novel	TRMT5	SPLICE_SITE	
chr14	64676125	64676125	Heterozygous	A	G	Novel	SLC38A6	UPSTREAM	
chr14	64676125	64676125	Heterozygous	A	G	Novel	SYNE2	INTRONIC	
chr14	64676125	64676125	Heterozygous	A	G	Novel	SYNE2	INTRONIC	
chr14	65472875	65472875	Heterozygous	T	-	Novel	FNTB	INTRONIC	
chr14	65472875	65472875	Heterozygous	T	-	Novel	FNTB	INTRONIC	
chr14	65472875	65472875	Heterozygous	T	-	Novel	MAX	DOWNSTREAM	
chr14	65733333	65733333	Heterozygous	-	T	Novel		GENIC	
chr14	65733792	65733792	Heterozygous	T	C	Novel		GENIC	
chr14	65733794	65733794	Heterozygous	T	G	Novel		GENIC	
chr14	66135968	66135968	Heterozygous	C	T	Novel	FUT8	SYNONYMOUS_CODING	
chr14	66135968	66135968	Heterozygous	C	T	Novel	FUT8	SYNONYMOUS_CODING	
chr14	66135968	66135968	Heterozygous	C	T	Novel	FUT8	SYNONYMOUS_CODING	
chr14	66135968	66135968	Heterozygous	C	T	Novel	FUT8	UPSTREAM	
chr14	66135968	66135968	Heterozygous	C	T	Novel	FUT8	SYNONYMOUS_CODING	
chr14	66188404	66188404	Heterozygous	G	A	Novel	FUT8	INTRONIC	
chr14	66188404	66188404	Heterozygous	G	A	Novel	FUT8	INTRONIC	
chr14	66188404	66188404	Heterozygous	G	A	Novel	FUT8	INTRONIC	
chr14	66188404	66188404	Heterozygous	G	A	Novel	FUT8	INTRONIC	
chr14	66188404	66188404	Heterozygous	G	A	Novel	FUT8	INTRONIC	
chr14	67859398	67859398	Heterozygous	G	C	Novel	PLEK2	NON_SYNONYIS->C	
chr14	68028616	68028616	Heterozygous	G	-	Novel	PLEKHH1	INTRONIC	
chr14	68129140	68129140	Heterozygous	T	C	Novel	VTT1B	INTRONIC	
chr14	68944357	68944357	Heterozygous	T	C	Novel	RAD51L1	SPLICE_SITE	
chr14	68944357	68944357	Heterozygous	T	C	Novel	RAD51L1	INTRONIC	
chr14	68944357	68944357	Heterozygous	T	C	Novel	RAD51L1	INTRONIC	
chr14	69376539	69376539	Heterozygous	C	G	Novel	ACTN1	INTRONIC	
chr14	69376539	69376539	Heterozygous	C	G	Novel			

Chromosome	Start	End	Zygosity	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr15	42880144	42880144	Heterozygous	C	G	Novel	CAPN3	INTRONIC	
chr15	42880144	42880144	Heterozygous	C	G	Novel	CAPN3	INTRONIC	
chr15	42880144	42880144	Heterozygous	C	G	Novel	CAPN3	INTRONIC	
chr15	42880144	42880144	Heterozygous	C	G	Novel	CAPN3	INTRONIC	
chr15	43501440	43501440	Heterozygous	C	G	Novel	EPB42	INTRONIC	
chr15	43501440	43501440	Heterozygous	C	G	Novel	EPB42	INTRONIC	
chr15	43552372	43552372	Heterozygous	C	T	Novel	TGM5	NON_SYNONYMIC->Y	
chr15	43552372	43552372	Heterozygous	C	T	Novel	TGM5	INTRONIC	
chr15	44080615	44080615	Heterozygous	C	T	Novel	PDIA3	INTRONIC	
chr15	44090252	44090252	Heterozygous	T	-	Novel	SERF2	DOWNSTREAM	
chr15	44090252	44090252	Heterozygous	T	-	Novel	SERINC4	INTRONIC	
chr15	44097202	44097202	Heterozygous	G	A	Novel	MFAP1	3PRIME_UTR	
chr15	44858159	44858159	Heterozygous	T	C	Novel	SPG11	NON_SYNONYMIC->V	
chr15	44858159	44858159	Heterozygous	T	C	Novel	SPG11	NON_SYNONYMIC->V	
chr15	45848231	45848231	Heterozygous	T	-	Novel	C15orf21	EXONIC	
chr15	48522754	48522754	Heterozygous	A	C	Novel	SLC12A1	INTRONIC	
chr15	48522754	48522754	Heterozygous	A	C	Novel	SLC12A1	INTRONIC	
chr15	49160089	49160089	Heterozygous	G	C	Novel	SHC4	INTRONIC	
chr15	50226457	50226457	Heterozygous	C	A	Novel	ATP8B4	INTRONIC	
chr15	50581870	50581870	Heterozygous	A	-	Novel	GABPB1	SPLICE_SITE	
chr15	50581870	50581870	Heterozygous	A	-	Novel	GABPB1	SPLICE_SITE	
chr15	50581870	50581870	Heterozygous	A	-	Novel	GABPB1	SPLICE_SITE	
chr15	50581870	50581870	Heterozygous	A	-	Novel	GABPB1	SPLICE_SITE	
chr15	50822216	50822216	Heterozygous	-	TT	Novel	USP50	INTRONIC	
chr15	50928524	50928524	Heterozygous	AAAT	---	Novel	TRPM7	INTRONIC	
chr15	52230440	52230440	Heterozygous	T	A	Novel	LEO1	SYNONYMOUS_CODING	
chr15	53178237	53178237	Heterozygous	C	G	Novel		GENIC	
chr15	53815528	53815528	Heterozygous	AG	-	Novel	WDR72	INTRONIC	
chr15	54307813	54307813	Heterozygous	T	C	Novel	UNC13G	NON_SYNONYMIC->H	
chr15	55790401	55790401	Heterozygous	T	C	Novel	DYX1C1	SPLICE_SITE	
chr15	55790401	55790401	Heterozygous	T	C	Novel	DYX1C1	SPLICE_SITE	
chr15	55790401	55790401	Heterozygous	T	C	Novel	DYX1C1	SPLICE_SITE	
chr15	58357691	58357691	Heterozygous	A	G	Novel	ALDH1A2	INTRONIC	
chr15	58357691	58357691	Heterozygous	A	G	Novel	ALDH1A2	INTRONIC	
chr15	59094495	59094495	Heterozygous	T	-	Novel	FAM63B	INTRONIC	
chr15	59094495	59094495	Heterozygous	T	-	Novel	FAM63B	INTRONIC	
chr15	62967564	62967564	Heterozygous	T	G	Novel	TLN2	INTRONIC	
chr15	63017267	63017267	Heterozygous	C	G	Novel	TLN2	SYNONYMOUS_CODING	
chr15	63030360	63030360	Heterozygous	A	G	Novel	TLN2	INTRONIC	
chr15	63032723	63032723	Heterozygous	A	G	Novel	TLN2	INTRONIC	
chr15	63040517	63040517	Heterozygous	A	-	Novel	TLN2	INTRONIC	
chr15	63638695	63638698	Heterozygous	CATA	---G	Novel	CA12	INTRONIC	
chr15	63638695	63638698	Heterozygous	CATA	---G	Novel	CA12	INTRONIC	
chr15	63950984	63950985	Heterozygous	-	A	Novel	HERC1	INTRONIC	
chr15	64011188	64011188	Heterozygous	T	C	Novel	HERC1	INTRONIC	
chr15	64496612	64496612	Heterozygous	A	-	Novel	CSNK1G1	INTRONIC	
chr15	64972781	64972782	Heterozygous	TG	-	Novel	ZNF809	INTRONIC	
chr15	65824472	65824472	Heterozygous	C	G	Novel	IGDCC3	INTRONIC	
chr15	65882489	65882489	Heterozygous	C	T	Novel	IGDCC4	SPLICE_SITE	
chr15	65884422	65884423	Heterozygous	-	C	Novel	IGDCC4	INTRONIC	
chr15	65874388	65874389	Heterozygous	-	A	Novel	C15orf44	INTRONIC	
chr15	65874388	65874389	Heterozygous	-	A	Novel	C15orf44	INTRONIC	
chr15	66833407	66833407	Heterozygous	C	-	Novel	TIPIN	INTRONIC	
chr15	66856980	66856980	Heterozygous	C	T	Novel	LC7L	INTRONIC	
chr15	69007719	69007719	Heterozygous	C	A	Novel		GENIC	
chr15	69373215	69373215	Heterozygous	G	T	Novel	NGRNA00277	EXONIC	
chr15	69709835	69709836	Heterozygous	----	TGAA	Novel	KIF23	INTRONIC	
chr15	69709835	69709836	Heterozygous	----	TGAA	Novel	KIF23	INTRONIC	
chr15	72231108	72231108	Homozygous	A	-	Novel	MYO9A	INTRONIC	
chr15	73542108	73542108	Heterozygous	T	C	Novel	NEO1	INTRONIC	
chr15	73542108	73542108	Heterozygous	T	C	Novel	NEO1	INTRONIC	
chr15	73542108	73542108	Heterozygous	T	C	Novel	NEO1	INTRONIC	
chr15	73575430	73575430	Heterozygous	C	T	Novel	NEO1	NON_SYNONYMIC->C	
chr15	73575430	73575430	Heterozygous	C	T	Novel	NEO1	NON_SYNONYMIC->C	
chr15	73575430	73575430	Heterozygous	C	T	Novel	NEO1	NON_SYNONYMIC->C	
chr15	74426144	74426144	Heterozygous	C	T	Novel	ISLR2	NON_SYNONYMIC->V	
chr15	74426144	74426144	Heterozygous	C	T	Novel	ISLR2	NON_SYNONYMIC->V	
chr15	74426144	74426144	Heterozygous	C	T	Novel	ISLR2	NON_SYNONYMIC->V	
chr15	74426144	74426144	Heterozygous	C	T	Novel	ISLR2	NON_SYNONYMIC->V	
chr15	74560828	74560829	Heterozygous	-	G	Novel	CCDC33	INTRONIC	
chr15	74836226	74836226	Heterozygous	C	T	Novel	ARID3B	5PRIME_UTR	
chr15	75649115	75649115	Homozygous	T	C	Novel	NEL1	DOWNSTREAM	
chr15	75649115	75649115	Homozygous	T	C	Novel	MAN2C1	INTRONIC	
chr15	76248134	76248134	Heterozygous	A	-	Novel	NRG4	INTRONIC	
chr15	78233938	78233938	Heterozygous	G	-	Novel		GENIC	
chr15	78458609	78458609	Heterozygous	G	A	Novel	IDH3A	NON_SYNONYMIC->K	

Chromosome	Start	End	Zygosity	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr15	78928022	78928022	Heterozygous	G	A	Novel	CHRNB4	INTRONIC	
chr15	79058980	79058980	Heterozygous	T	C	Novel	ADAMTS7	SYNONYMOUS_CODING	
chr15	80414038	80414038	Heterozygous	T	C	Novel	ZFAND6	INTRONIC	
chr15	84442240	84442241	Heterozygous	-	-T	Novel	ADAMTSL3	INTRONIC	
chr15	84442240	84442241	Heterozygous	-	TT	Novel	ADAMTSL3	INTRONIC	
chr15	85656767	85656767	Heterozygous	G	A	Novel	PDE8A	INTRONIC	
chr15	85656767	85656767	Heterozygous	G	A	Novel	PDE8A	INTRONIC	
chr15	88428899	88428899	Heterozygous	-	A	Novel	NTRK3	INTRONIC	
chr15	88428899	88428899	Heterozygous	-	A	Novel	NTRK3	INTRONIC	
chr15	91549253	91549253	Heterozygous	C	T	Novel	VPS33B	NON_SYNONYMIC->I	
chr15	93510829	93510829	Heterozygous	T	-	Novel	CHD2	INTRONIC	
chr15	98513087	98513087	Heterozygous	T	-	Novel	ARRDC4	INTRONIC	
chr15	10033119	10033119	Homozygous	C	T	Novel	C15orf51	EXONIC	
chr15	100339833	100339833	Homozygous	G	C	Novel	C15orf51	SPLICE_SITE	
chr15	100340175	100340176	Heterozygous	TG	AT	Novel	C15orf51	EXONIC	
chr15	100638407	100638407	Heterozygous	T	C	Novel	ADAMTS17	INTRONIC	
chr18	541087	541087	Heterozygous	C	T	Novel	RAB11FIP3	INTRONIC	
chr18	541087	541087	Heterozygous	C	T	Novel	RAB11FIP3	INTRONIC	
chr18	541082	541082	Heterozygous	A	G	Novel	RAB11FIP3	INTRONIC	
chr18	541082	541082	Heterozygous	A	G	Novel	RAB11FIP3	INTRONIC	
chr18	830805	830805	Heterozygous	A	G	Novel	MSLN1	INTRONIC	
chr18	838914	838914	Heterozygous	T	G	Novel	RPUSD1	NON_SYNONYMIC->Q	
chr18	838914	838914	Heterozygous	T	G	Novel	CHTF18	INTRONIC	
chr18	839987	839987	Heterozygous	C	T	Novel	RPUSD1	UPSTREAM	
chr18	839987	839987	Heterozygous	C	T	Novel	CHTF18	INTRONIC	
chr18	1291182	1291182	Heterozygous	C	T	Novel	TPSAB1	SYNONYMOUS_CODING	
chr18	1573895	1573895	Heterozygous	C	A	Novel	IFT140	SYNONYMOUS_CODING	
chr18	1842461	1842461	Heterozygous	C	T	Novel	IFT140	SPLICE_SITE	
chr18	2098768	2098768	Heterozygous	C	G	Novel	NTHL1	UPSTREAM	
chr18	2098768	2098768	Heterozygous	C	G	Novel	TS2C2	INTRONIC	
chr18	2098768	2098768	Heterozygous	C	G	Novel	TS2C2	INTRONIC	
chr18	2098768	2098768	Heterozygous	C	G	Novel	TS2C2	INTRONIC	
chr18	2141958	2141958	Heterozygous	C	T	Novel	PKD1	INTRONIC	
chr18	2141958	2141958	Heterozygous	C	T	Novel	PKD1	INTRONIC	
chr18	2153531	2153531	Heterozygous	T	C	Novel	PKD1	NON_SYNONYMIC->A	
chr18	2153531	2153531	Heterozygous	T	C	Novel	PKD1	NON_SYNONYMIC->A	
chr18	2160358	2160358	Heterozygous	C	T	Novel	PKD1	NON_SYNONYMIC->M	
chr18	2160358	2160358	Heterozygous	C	T	Novel	PKD1	NON_SYNONYMIC->M	
chr18	3434820	3434820	Heterozygous	C	T	Novel	ZNF434	SYNONYMOUS_CODING	
chr18	3576345	3576345	Heterozygous	A	A	Novel	CLUAP1	INTRONIC	
chr18	3576345	3576345	Heterozygous	A	A	Novel	CLUAP1	INTRONIC	
chr18	4256756	4256756	Heterozygous	C	-	Novel	SLR1	INTRONIC	
chr18	4700319	4700319	Heterozygous	A	-	Novel	MGRN1	INTRONIC	
chr18	4700319	4700319	Heterozygous	A	-	Novel	MGRN1	INTRONIC	
chr18	4700319	4700319	Heterozygous	A	-	Novel	MGRN1	INTRONIC	
chr18	4700319	4700319	Heterozygous	A	-	Novel	MGRN1	INTRONIC	
chr18	4752158	4752158	Heterozygous	C	A	Novel	ANKS3	SYNONYMOUS_CODING	
chr18	4934597	4934597	Heterozygous	C	T	Novel	PPL	SYNONYMOUS_CODING	
chr18	5143803	5143803	Heterozygous	C	T	Novel	FAM88A	INTRONIC	
chr18	5143803	5143803	Heterozygous	C	T	Novel	FAM88A	INTRONIC	
chr18	8952382	8952382	Heterozygous	A	C	Novel	CARHSP1	INTRONIC	
chr18	8952382	8952382	Heterozygous	A	C	Novel	CARHSP1	INTRONIC	
chr18	8991118	8991118	Heterozygous	-	A	Novel	USP7	INTRONIC	
chr18	10576102	10576102	Heterozygous	C	T	Novel	ATF7IP2	NON_SYNONYMIC->M	
chr18	10626060	10626060	Heterozygous	T	-	Novel	EMP2	3PRIME_UTR	
chr18	10934475	10934475	Heterozygous	C	C	Novel		GENIC	
chr18	11133702	11133702	Heterozygous	C	G	Novel	CLEC16A	SYNONYMOUS_CODING	
chr18	11370108	11370108	Heterozygous	A	A	Novel	PRM2	SYNONYMOUS_CODING	
chr18	11444455	11444455	Homozygous	A	-	Novel	C16orf75	INTRONIC	
chr18	11870553	11870553	Heterozygous	T	C	Novel	ZC3H7A	INTRONIC	
chr18	11935480	11935481	Heterozygous	TG	-	Novel	RSL1D1	INTRONIC	
chr18	12021486	12021486	Heterozygous	C	A	Novel		GENIC	
chr18	15045766	15045766	Heterozygous	C	T	Novel	NPIP	NON_SYNONYMIC->F	
chr18	15083383	15083383	Heterozygous	G	A	Novel	PDXDC1	INTRONIC	
chr18	15457642	15457642	Heterozygous						

Chromosome	Start	End	Zygosity	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr16	21901504	21901504	Heterozygous	A	T	Novel	GENIC		
chr16	21902060	21902061	Heterozygous	-	A	Novel	GENIC		
chr16	22545865	22545865	Heterozygous	G	C	Novel	LOC1001322:NON_SYNONYI	A->P	
chr16	22545897	22545898	Heterozygous	GA	GC	Novel	LOC1001322:NON_SYNONYI	KT->KP	
chr16	22545897	22545898	Heterozygous	GA	TA	Novel	LOC1001322:NON_SYNONYI	KT->NT	
chr16	24820824	24820824	Heterozygous	A	-	Novel	TNRC6A	INTRONIC	
chr16	24886887	24886887	Homozygous	G	-	Novel	SLCS5A11	INTRONIC	
chr16	28998098	28998098	Heterozygous	G	A	Novel	LAT	INTRONIC	
chr16	28998098	28998098	Heterozygous	G	A	Novel	LAT	INTRONIC	
chr16	28998098	28998098	Heterozygous	G	A	Novel	LAT	INTRONIC	
chr16	28998098	28998098	Heterozygous	G	A	Novel	LAT	INTRONIC	
chr16	29113323	29113323	Heterozygous	C	A	Novel	RRN3P2	EXONIC	
chr16	29387272	29387272	Heterozygous	C	T	Novel	RUNDCC2C	EXONIC	
chr16	30001081	30001081	Heterozygous	G	A	Novel	TAOK2	SYNONYMOUS_CODING	
chr16	30001081	30001081	Heterozygous	G	A	Novel	TAOK2	DOWNSTREAM	
chr16	30004603	30004603	Heterozygous	T	G	Novel	TAOK2	DOWNSTREAM	
chr16	30004603	30004603	Heterozygous	T	G	Novel	HIRIP3	NON_SYNONYI	E->D
chr16	30005505	30005505	Heterozygous	T	C	Novel	TAOK2	DOWNSTREAM	
chr16	30005505	30005505	Heterozygous	T	C	Novel	HIRIP3	NON_SYNONYI	T->A
chr16	30731430	30731430	Heterozygous	C	C	Novel	SRCAP	INTRONIC	
chr16	31102595	31102595	Heterozygous	C	G	Novel	VKORC1	NON_SYNONYI	C->S
chr16	31102595	31102595	Heterozygous	C	G	Novel	VKORC1	NON_SYNONYI	V->L
chr16	32437070	32437071	Heterozygous	-	A	Novel	GENIC		
chr16	33710045	33710045	Heterozygous	G	T	Novel	GPT2	5PRIME_UTR	
chr16	48931607	48931607	Heterozygous	C	T	Novel	GPT2	SYNONYMOUS_CODING	
chr16	48931607	48931607	Heterozygous	C	T	Novel	PHKB	SYNONYMOUS_CODING	
chr16	47703131	47703131	Heterozygous	T	G	Novel	PHKB	SYNONYMOUS_CODING	
chr16	47703131	47703131	Heterozygous	T	G	Novel	BRD7	INTRONIC	
chr16	50383879	50383879	Heterozygous	T	C	Novel	BRD7	INTRONIC	
chr16	50383879	50383879	Heterozygous	T	C	Novel	CHD9	INTRONIC	
chr16	53289032	53289032	Heterozygous	C	-	Novel	CHD9	INTRONIC	
chr16	53325597	53325597	Heterozygous	C	A	Novel	RBL2	NON_SYNONYI	N->I
chr16	53476621	53476621	Heterozygous	A	G	Novel	RBL2	NON_SYNONYI	N->I
chr16	53498223	53498223	Heterozygous	A	T	Novel	OGFOD1	INTRONIC	
chr16	56504721	56504721	Heterozygous	A	A	Novel	MT52	INTRONIC	
chr16	56535194	56535194	Heterozygous	A	-	Novel	BT4	INTRONIC	
chr16	56602672	56602672	Heterozygous	T	G	Novel	MT4	INTRONIC	
chr16	56855550	56855550	Heterozygous	C	-	Novel	NUP93	INTRONIC	
chr16	56870627	56870627	Heterozygous	A	C	Novel	NUP93	NON_SYNONYI	K->Q
chr16	57080877	57080877	Heterozygous	G	A	Novel	NLRC5	SYNONYMOUS_CODING	
chr16	57788739	57788739	Heterozygous	C	G	Novel	KATNB1	INTRONIC	
chr16	58030595	58030595	Heterozygous	C	T	Novel	ZNF319	SYNONYMOUS_CODING	
chr16	58620361	58620361	Homozygous	T	G	Novel	CNOT1	INTRONIC	
chr16	58620361	58620361	Homozygous	T	G	Novel	CNOT1	INTRONIC	
chr16	58701316	58701316	Heterozygous	G	A	Novel	SLC38A7	SYNONYMOUS_CODING	
chr16	58743518	58743518	Heterozygous	C	T	Novel	GOT2	INTRONIC	
chr16	66432496	66432496	Heterozygous	G	A	Novel	CDH5	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	66605008	66605009	Heterozygous	-	G	Novel	CMTM1	INTRONIC	
chr16	67285154	67285154	Heterozygous	G	C	Novel	TMEM208	DOWNSTREAM	
chr16	67285154	67285154	Heterozygous	G	C	Novel	FHOD1	SYNONYMOUS_CODING	
chr16	67862706	67862706	Heterozygous	G	C	Novel	TSNAXIP1	DOWNSTREAM	
chr16	67862706	67862706	Heterozygous	G	C	Novel	CENPT	NON_SYNONYI	P->A
chr16	68112725	68112725	Heterozygous	C	T	Novel	ODH2L	NON_SYNONYI	R->W
chr16	68842784	68842784	Heterozygous	G	A	Novel	CDH1	INTRONIC	
chr16	69143279	69143279	Heterozygous	C	T	Novel	HAS3	INTRONIC	
chr16	69143279	69143279	Heterozygous	C	T	Novel	HAS3	INTRONIC	
chr16	69352948	69352948	Heterozygous	C	A	Novel	VPS4A	INTRONIC	
chr16	70010610	70010610	Heterozygous	C	T	Novel	PDXDC2P	EXONIC	
chr16	70182333	70182333	Heterozygous	G	C	Novel	PDPFR	INTRONIC	
chr16	70299590	70299591	Heterozygous	-	A	Novel	AARS	INTRONIC	
chr16	70367494	70367494	Heterozygous	A	C	Novel	DDX19B	3PRIME_UTR	
chr16	70367494	70367494	Heterozygous	A	C	Novel	DDX19B	3PRIME_UTR	
chr16	70367494	70367494	Heterozygous	A	C	Novel	DDX19B	3PRIME_UTR	
chr16	70389914	70389914	Heterozygous	C	G	Novel	DDX19A	INTRONIC	
chr16	70405898	70405898	Heterozygous	A	C	Novel	DDX19A	3PRIME_UTR	
chr16	70694048	70694048	Heterozygous	G	C	Novel	MTSSL1	DOWNSTREAM	
chr16	70698492	70698492	Heterozygous	C	T	Novel	MTSSL1L	INTRONIC	
chr16	71799358	71799358	Heterozygous	A	G	Novel	AP1G1	INTRONIC	
chr16	71799358	71799358	Heterozygous	A	G	Novel	AP1G1	INTRONIC	
chr16	72130772	72130772	Heterozygous	C	T	Novel	DHX38	SYNONYMOUS_CODING	

Chromosome	Start	End	Zygosity	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr16	72821594	72821596	Heterozygous	GCC	---	Novel	ZFH3	NON_SYNONYMOUS_CODING	
chr16	72821594	72821596	Heterozygous	GCC	---	Novel	ZFH3	NON_SYNONYMOUS_CODING	
chr16	72821615	72821615	Heterozygous	G	A	Novel	ZFH3	SYNONYMOUS_CODING	
chr16	72821615	72821615	Heterozygous	G	A	Novel	ZFH3	SYNONYMOUS_CODING	
chr16	75874253	75874254	Heterozygous	AA	---	Novel	KARS	SPICE_SITE	
chr16	75874253	75874254	Heterozygous	AA	-A	Novel	KARS	SPICE_SITE	
chr16	75874253	75874254	Heterozygous	AA	---	Novel	KARS	SPICE_SITE	
chr16	75874253	75874254	Heterozygous	AA	-A	Novel	KARS	SPICE_SITE	
chr16	75875514	75875514	Heterozygous	T	C	Novel	KARS	NON_SYNONYI	N->S
chr16	75875514	75875514	Heterozygous	T	C	Novel	KARS	NON_SYNONYI	N->S
chr16	77225217	77225218	Heterozygous	-	G	Novel	MON1B	INTRONIC	
chr16	78064752	78064752	Heterozygous	G	C	Novel	CLEG3A	3PRIME_UTR	
chr16	81050938	81050938	Heterozygous	A	G	Novel	CENPN	SYNONYMOUS_CODING	
chr16	81050938	81050938	Heterozygous	A	G	Novel	CENPN	SYNONYMOUS_CODING	
chr16	81050938	81050938	Heterozygous	A	G	Novel	CENPN	SYNONYMOUS_CODING	
chr16	81082157	81082157	Heterozygous	T	C	Novel	CENPN	INTRONIC	
chr16	81082157	81082157	Heterozygous	T	C	Novel	CENPN	INTRONIC	
chr16	81190430	81190430	Heterozygous	C	T	Novel	PKD1L2	INTRONIC	
chr16	81192655	81192655	Heterozygous	G	A	Novel	PKD1L2	INTRONIC	
chr16	81222448	81222448	Heterozygous	A	-	Novel	PKD1L2	INTRONIC	
chr16	81222448	81222448	Heterozygous	A	-	Novel	PKD1L2	INTRONIC	
chr16	81293507	81293507	Heterozygous	G	C	Novel	BCMO1	INTRONIC	
chr16	83711779	83711779	Heterozygous	T	G	Novel	CDH13	INTRONIC	
chr16	84209590	84209590	Heterozygous	G	A	Novel	LRRCS0	NON_SYNONYI	D->N
chr16	84209590	84209590	Heterozygous	G	A	Novel	TAFC1	DOWNSTREAM	
chr16	84209590	84209590	Heterozygous	G	A	Novel	TAFC1	DOWNSTREAM	
chr16	87457532	87457532	Heterozygous	T	A	Novel	ZCCHC14	INTRONIC	
chr16	88786904	88786904	Heterozygous	G	A	Novel	GENIC		
chr16	89576880	89576881	Heterozygous	TT	-T	Novel	SPG7	INTRONIC	
chr16	89576880	89576881	Heterozygous	TT	---	Novel	SPG7	INTRONIC	
chr16	89576880	89576881	Heterozygous	TT	-T	Novel	SPG7	INTRONIC	
chr16	89576880	89576881	Heterozygous	TT	---	Novel	SPG7	INTRONIC	
chr16	89629468	89629468	Heterozygous	G	T	Novel	RPL13	3PRIME_UTR	
chr16	89629468	89629468	Heterozygous	G	T	Novel	RPL13	3PRIME_UTR	
chr16	89629468	89629468	Heterozygous	G	T	Novel	SNORD68	DOWNSTREAM	
chr16	89838157	89838157	Heterozygous	C	T	Novel	FANCA	NON_SYNONYI	D->N
chr16	90029868	90029868	Heterozygous	G	T	Novel	DEF8	INTRONIC	
chr17	293292	293292	Heterozygous	G	C	Novel	FAM101B	SPICE_SITE	
chr17	602613	602613	Heterozygous	G	A	Novel	VPS53	NON_SYNONYI	A->V
chr17	602613	602613	Heterozygous	G	A	Novel	VPS53	NON_SYNONYI	A->V
chr17	686590	686590	Heterozygous	-	T	Novel	GLOD4	UPSTREAM	
chr17	686590	686590	Heterozygous	-	T	Novel	RNMTL1	INTRONIC	
chr17	1248666	1248666	Heterozygous	G	A	Novel	YWHAE	3PRIME_UTR	
chr17	1248666	1248666	Heterozygous	G	A	Novel	YWHAE	EXONIC	
chr17	1264612	1264612	Heterozygous	A	-	Novel	YWHAE	INTRONIC	
chr17	1264612	1264612	Heterozygous	A	-	Novel	YWHAE	INTRONIC	
chr17	1326754	1326755	Heterozygous	-	A	Novel	CRK	3PRIME_UTR	
chr17	1326754	1326755	Heterozygous	-	A	Novel	CRK	3PRIME_UTR	
chr17	1486521	1486521	Heterozygous	G	A	Novel	SLC43A2	NON_SYNONYI	L->F
chr17	1959577	1959577	Heterozygous	C	G	Novel	HIC1	INTRONIC	
chr17	1959577	1959577	Heterozygous	C	G	Novel	HIC1	UPSTREAM	
chr17	2239725	2239725	Heterozygous	G	A	Novel	TSR1	5PRIME_UTR	
chr17	2239725	2239725	Heterozygous	G	A	Novel	SGSM2	UPSTREAM	
chr17	2239725	2239725	Heterozygous	G	A	Novel	SGSM2	UPSTREAM	
chr17	2239725	2239725	Heterozygous	G	A	Novel	SGSM2	UPSTREAM	
chr17	2281267	2281267	Heterozygous	A	G	Novel	SGSM2	INTRONIC	
chr17	2281267	2281267	Heterozygous	A	G	Novel	SGSM2	INTRONIC	
chr17	2867437	2867437	Heterozygous	C	T	Novel	RAP1GAP2	INTRONIC	
chr17	2867437	2867437	Heterozygous	C	T	Novel	RAP1GAP2	INTRONIC	
chr17	3030822	3030822	Heterozygous	G	A	Novel	OR1G1	SYNONYMOUS_CODING	
chr17	3301535	3301535	Heterozygous	G	A	Novel	OR1E1	NON_SYNONYI	T->M
chr17	3962428	3962428	Heterozygous	-	T	Novel	ZZF1	INTRONIC	
chr17	4439731	44397							

Chromosome	Start	End	Zygosity	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr17	5433769	5433769	Heterozygous	C	A	Novel	NLRP1	INTRONIC	
chr17	5433769	5433769	Heterozygous	C	A	Novel	NLRP1	INTRONIC	
chr17	5433769	5433769	Heterozygous	C	A	Novel	NLRP1	INTRONIC	
chr17	5433769	5433769	Heterozygous	C	A	Novel	NLRP1	INTRONIC	
chr17	5436343	5436343	Heterozygous	G	A	Novel	NLRP1	INTRONIC	
chr17	5436343	5436343	Heterozygous	G	A	Novel	NLRP1	INTRONIC	
chr17	5436343	5436343	Heterozygous	G	A	Novel	NLRP1	INTRONIC	
chr17	5436343	5436343	Heterozygous	G	A	Novel	NLRP1	INTRONIC	
chr17	5436343	5436343	Heterozygous	G	A	Novel	NLRP1	INTRONIC	
chr17	5436343	5436343	Heterozygous	G	A	Novel	NLRP1	INTRONIC	
chr17	6330227	6330227	Heterozygous	T	C	Novel	AIP1L	NON_SYNONYMI->V	
chr17	6330227	6330227	Heterozygous	T	C	Novel	AIP1L	NON_SYNONYMI->V	
chr17	6330227	6330227	Heterozygous	T	C	Novel	AIP1L	NON_SYNONYMI->V	
chr17	6540267	6540267	Heterozygous	A	G	Novel	TXNDC17	SPLICE_SITE	
chr17	6540267	6540267	Heterozygous	A	G	Novel	TXNDC17	SPLICE_SITE	
chr17	6540267	6540267	Heterozygous	A	G	Novel	TXNDC17	SPLICE_SITE	
chr17	6901880	6901880	Heterozygous	G	A	Novel	ALOX12	SYNONYMOUS_CODING	
chr17	6903097	6903097	Heterozygous	G	A	Novel	BCL6B	SYNONYMOUS_CODING	
chr17	7106694	7106694	Heterozygous	G	-	Novel	DLG4	INTRONIC	
chr17	7106694	7106694	Heterozygous	G	-	Novel	DLG4	INTRONIC	
chr17	7125654	7125654	Heterozygous	T	G	Novel	ACADVL	INTRONIC	
chr17	7125654	7125654	Heterozygous	T	G	Novel	ACADVL	INTRONIC	
chr17	7129429	7129429	Heterozygous	G	A	Novel	ACADVL	DOWNSTREAM	
chr17	7129429	7129429	Heterozygous	G	A	Novel	ACADVL	DOWNSTREAM	
chr17	7129429	7129429	Heterozygous	G	A	Novel	DVL2	NON_SYNONYMI->S	
chr17	7211904	7211905	Heterozygous	-	C	Novel	EIF5A	INTRONIC	
chr17	7211904	7211905	Heterozygous	-	C	Novel	EIF5A	INTRONIC	
chr17	7211904	7211905	Heterozygous	-	C	Novel	EIF5A	INTRONIC	
chr17	7211904	7211905	Heterozygous	-	C	Novel	EIF5A	INTRONIC	
chr17	7259083	7259084	Heterozygous	-	G	Novel	KGDT11	DOWNSTREAM	
chr17	7259083	7259084	Heterozygous	-	G	Novel	TMEM95	INTRONIC	
chr17	7686653	7686653	Heterozygous	G	C	Novel	DNAH2	INTRONIC	
chr17	7671162	7671162	Heterozygous	G	C	Novel	DNAH2	INTRONIC	
chr17	7679352	7679352	Heterozygous	G	A	Novel	DNAH2	NON_SYNONYMI->D	
chr17	7721200	7721200	Heterozygous	C	-	Novel	DNAH2	INTRONIC	
chr17	7722127	7722127	Heterozygous	C	-	Novel	DNAH2	INTRONIC	
chr17	7722130	7722130	Heterozygous	G	C	Novel	DNAH2	INTRONIC	
chr17	7814668	7814668	Heterozygous	T	C	Novel	CHD3	INTRONIC	
chr17	7814668	7814668	Heterozygous	T	C	Novel	CHD3	INTRONIC	
chr17	7814668	7814668	Heterozygous	T	C	Novel	CHD3	INTRONIC	
chr17	7814668	7814668	Heterozygous	T	C	Novel	CHD3	INTRONIC	
chr17	7948185	7948185	Heterozygous	G	A	Novel	LOC284023	DOWNSTREAM	
chr17	7948185	7948185	Heterozygous	G	A	Novel	ALOX15B	NON_SYNONYMI->T	
chr17	7948185	7948185	Heterozygous	G	A	Novel	ALOX15B	NON_SYNONYMI->T	
chr17	7948185	7948185	Heterozygous	G	A	Novel	ALOX15B	NON_SYNONYMI->T	
chr17	8044256	8044256	Heterozygous	C	T	Novel	PER1	3PRIME_UTR	
chr17	8045029	8045029	Heterozygous	T	-	Novel	PER1	INTRONIC	
chr17	8387443	8387443	Heterozygous	G	A	Novel	MYH10	INTRONIC	
chr17	10367729	10367729	Heterozygous	T	A	Novel	MYH4	INTRONIC	
chr17	10398186	10398186	Heterozygous	G	A	Novel	MYH1	INTRONIC	
chr17	12799593	12799594	Heterozygous	AA	-	Novel	ARHGAP44	INTRONIC	
chr17	13977813	13977814	Heterozygous	---	TTTA	Novel	COX10	INTRONIC	
chr17	15343536	15343536	Heterozygous	A	T	Novel	CDRT4	NON_SYNONYMI->K	
chr17	15468766	15468766	Heterozygous	A	-	Novel	FAM18B2	UPSTREAM	
chr17	15468766	15468766	Heterozygous	A	-	Novel	FAM18B2	UPSTREAM	
chr17	15973418	15973418	Heterozygous	T	-	Novel	NGOR1	INTRONIC	
chr17	15973418	15973418	Heterozygous	T	-	Novel	NGOR1	INTRONIC	
chr17	16946210	16946210	Heterozygous	C	T	Novel	MPRIP	SYNONYMOUS_CODING	
chr17	16946210	16946210	Heterozygous	C	T	Novel	MPRIP	SYNONYMOUS_CODING	
chr17	17152364	17152364	Heterozygous	A	-	Novel	COPS3	INTRONIC	
chr17	17188053	17188054	Heterozygous	-	T	Novel	COPS3	INTRONIC	
chr17	17188367	17188367	Heterozygous	A	T	Novel	COPS3	INTRONIC	
chr17	17719368	17719368	Heterozygous	A	-	Novel	SREBF1	INTRONIC	
chr17	17719368	17719368	Heterozygous	A	-	Novel	SREBF1	INTRONIC	
chr17	18148858	18148858	Heterozygous	C	A	Novel	LLGL1	DOWNSTREAM	
chr17	18148858	18148858	Heterozygous	C	A	Novel	FLII	INTRONIC	
chr17	18394676	18394676	Heterozygous	T	C	Novel	LGALS9C	INTRONIC	
chr17	18513456	18513456	Heterozygous	G	A	Novel	CCDC144B	EXONIC	
chr17	19186967	19186967	Heterozygous	A	C	Novel	EPN2	NON_SYNONYMI->P	
chr17	19186967	19186967	Heterozygous	A	C	Novel	EPN2	NON_SYNONYMI->P	
chr17	19186967	19186967	Heterozygous	A	C	Novel	EPN2	INTRONIC	
chr17	19283802	19283802	Heterozygous	A	G	Novel	MAPK7	INTRONIC	
chr17	19283802	19283802	Heterozygous	A	G	Novel	MAPK7	INTRONIC	
chr17	19283802	19283802	Heterozygous	A	G	Novel	MAPK7	INTRONIC	
chr17	19283802	19283802	Heterozygous	A	G	Novel	MAPK7	INTRONIC	
chr17	19643616	19643616	Heterozygous	G	A	Novel	ALDH3A1	INTRONIC	
chr17	19643616	19643616	Heterozygous	G	A	Novel	ALDH3A1	INTRONIC	
chr17	19643616	19643616	Heterozygous	G	A	Novel	ALDH3A1	INTRONIC	
chr17	19850685	19850686	Heterozygous	-	T	Novel	AKAP10	INTRONIC	
chr17	20108027	20108027	Heterozygous	C	T	Novel	SPECC1	NON_SYNONYMI->M	

Chromosome	Start	End	Zygosity	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr17	20108027	20108027	Heterozygous	C	T	Novel	SPECC1	NON_SYNONYMI->M	
chr17	20108027	20108027	Heterozygous	C	T	Novel	SPECC1	NON_SYNONYMI->M	
chr17	20108027	20108027	Heterozygous	C	T	Novel	SPECC1	NON_SYNONYMI->M	
chr17	20108050	20108052	Heterozygous	GAG	---	Novel	SPECC1	NON_SYNONYMOUS_CODING	
chr17	20108050	20108052	Heterozygous	GAG	---	Novel	SPECC1	NON_SYNONYMOUS_CODING	
chr17	20108050	20108052	Heterozygous	GAG	---	Novel	SPECC1	NON_SYNONYMOUS_CODING	
chr17	20108050	20108052	Heterozygous	GAG	---	Novel	SPECC1	NON_SYNONYMOUS_CODING	
chr17	20332056	20332056	Homozygous	G	T	Novel	GENIC		
chr17	20332072	20332072	Homozygous	A	C	Novel	GENIC		
chr17	20420444	20420444	Heterozygous	G	C	Novel	GENIC		
chr17	20634664	20634664	Heterozygous	G	A	Novel	GENIC		
chr17	21319651	21319653	Heterozygous	GAG	---	Novel	KCNJ12	NON_SYNONYMOUS_CODING	
chr17	26911789	26911789	Heterozygous	C	T	Novel	SPAG5	INTRONIC	
chr17	27420017	27420018	Heterozygous	CA	AC	Novel	MYO18A	NON_SYNONYMI->G	
chr17	27420017	27420018	Heterozygous	CA	AC	Novel	MYO18A	NON_SYNONYMI->G	
chr17	27778596	27778596	Heterozygous	G	A	Novel	TAOK1	SYNONYMOUS_CODING	
chr17	27959264	27959264	Heterozygous	G	A	Novel	SSH2	NON_SYNONYMI->V	
chr17	28754635	28754635	Heterozygous	T	G	Novel	CPD	INTRONIC	
chr17	28898253	28898253	Heterozygous	C	T	Novel	GENIC		
chr17	29848404	29848404	Heterozygous	T	C	Novel	RAB11FIP4	INTRONIC	
chr17	29850895	29850895	Heterozygous	C	T	Novel	RAB11FIP4	INTRONIC	
chr17	30348180	30348180	Heterozygous	T	C	Novel	LRRG37B	5PRIME_UTR	
chr17	30470028	30470028	Heterozygous	-	C	Novel	RHOT1	INTRONIC	
chr17	30470028	30470028	Heterozygous	-	C	Novel	RHOT1	INTRONIC	
chr17	30470028	30470028	Heterozygous	-	C	Novel	RHOT1	INTRONIC	
chr17	33318215	33318215	Heterozygous	T	C	Novel	LIQ3	INTRONIC	
chr17	33318215	33318215	Heterozygous	T	C	Novel	LIQ3	INTRONIC	
chr17	33329653	33329653	Heterozygous	C	T	Novel	LIQ3	INTRONIC	
chr17	33329653	33329653	Heterozygous	C	T	Novel	LIQ3	INTRONIC	
chr17	33331358	33331358	Heterozygous	G	A	Novel	LIQ3	NON_SYNONYMI->H	
chr17	33331358	33331358	Heterozygous	G	A	Novel	LIQ3	DOWNSTREAM	
chr17	33445416	33445417	Heterozygous	-	A	Novel	RAD51L3	INTRONIC	
chr17	33445416	33445417	Heterozygous	-	A	Novel	RAD51L3	INTRONIC	
chr17	33445416	33445417	Heterozygous	-	A	Novel	RAD51L3	INTRONIC	
chr17	33445416	33445417	Heterozygous	-	A	Novel	RAD51L3	INTRONIC	
chr17	33806842	33806842	Heterozygous	G	-	Novel	SLFN1L2	FRAMESHIFT_CODING	
chr17	34539815	34539815	Heterozygous	G	C	Novel	CCL4L1	INTRONIC	
chr17	35980638	35980638	Heterozygous	C	A	Novel	SYNRG	INTRONIC	
chr17	35980638	35980638	Heterozygous	C	A	Novel	SYNRG	INTRONIC	
chr17	35980638	35980638	Heterozygous	C	A	Novel	SYNRG	INTRONIC	
chr17	35980638	35980638	Heterozygous	C	A	Novel	SYNRG	INTRONIC	
chr17	35980638	35980638	Heterozygous	C	A	Novel	SYNRG	INTRONIC	
chr17	37034514	37034514	Heterozygous	C	T	Novel	LASP1	INTRONIC	
chr17	37657460	37657461	Heterozygous	-	T	Novel	CDK12	INTRONIC	
chr17	37657460	37657461	Heterozygous	-	T	Novel	CDK12	INTRONIC	
chr17	38935899	38935899	Heterozygous	G	C	Novel	KRT27	INTRONIC	
chr17	39135030	39135030	Heterozygous	A	T	Novel	GENIC		
chr17	39137137	39137137	Heterozygous	A	G	Novel	GENIC		
chr17	39183254	39183254	Heterozygous	T	C	Novel	KRTAP1-5	NON_SYNONYMI->A	
chr17	39186052	39186052	Heterozygous	G	A	Novel	GENIC		
chr17	39189195	39189196	Heterozygous	CA	TA	Novel	GENIC		
chr17	39190939	39190939	Heterozygous	CA	TA	Novel	GENIC		
chr17	39197638	39197638	Heterozygous	A	A	Novel	KRTAP1-3	SYNONYMOUS_CODING	
chr17	39240627	39240627	Heterozygous	T	C	Novel	KRTAP1-1	SYNONYMOUS_CODING	
chr17	39254318	39254318	Heterozygous	C	T	Novel	GENIC		
chr17	39254335	39254336	Heterozygous	-	T	Novel	GENIC		
chr17	39258344	39258344	Heterozygous	A	G	Novel	GENIC		
chr17	39274491	39274491	Heterozygous	C	T	Novel	KRTAP4-11	NON_SYNONYMI->H	
chr17	39279841	39279841	Heterozygous	G	A	Novel	KRTAP4-12	SYNONYMOUS_CODING	
chr17	39388811	39388811	Heterozygous	C	A	Novel	KRTAP9-3	NON_SYNONYMI->K	
chr17	39537475	39537475	Heterozygous	C	T	Novel	KRT34	INTRONIC	
chr17	39								

Chromosome	Start	End	Zygoty	Reference	SNP Call	dSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr21	18931473	18931473	Heterozygous A		-	Novel	CXADR	INTRONIC	
chr21	19653343	19653343	Heterozygous T		-	Novel	TMPRSS15	INTRONIC	
chr21	33873873	33873873	Heterozygous G		C	Novel	C21orf63	INTRONIC	
chr21	33949071	33949071	Heterozygous C		G	Novel	TOP10L	3PRIME_UTR	
chr21	34050940	34050940	Heterozygous A		-	Novel	SYNJ1	INTRONIC	
chr21	34050940	34050940	Heterozygous A		-	Novel	SYNJ1	INTRONIC	
chr21	34050940	34050940	Heterozygous A		-	Novel	SYNJ1	INTRONIC	
chr21	34050940	34050940	Heterozygous A		-	Novel	SYNJ1	INTRONIC	
chr21	34050988	34050988	Heterozygous C		G	Novel	SYNJ1	NON_SYNONYMI>P	
chr21	34050988	34050988	Heterozygous C		G	Novel	SYNJ1	NON_SYNONYMI>P	
chr21	34050988	34050988	Heterozygous C		G	Novel	SYNJ1	NON_SYNONYMI>P	
chr21	34050988	34050988	Heterozygous C		G	Novel	SYNJ1	NON_SYNONYMI>P	
chr21	34066492	34066492	Heterozygous T		C	Novel	SYNJ1	INTRONIC	
chr21	34066492	34066492	Heterozygous T		C	Novel	SYNJ1	INTRONIC	
chr21	34066492	34066492	Heterozygous T		C	Novel	SYNJ1	INTRONIC	
chr21	34066492	34066492	Heterozygous T		C	Novel	SYNJ1	INTRONIC	
chr21	34127440	34127440	Heterozygous A		-	Novel	GGFC1	INTRONIC	
chr21	34127440	34127440	Heterozygous A		-	Novel	GGFC1	INTRONIC	
chr21	34127440	34127440	Heterozygous A		-	Novel	GGFC1	INTRONIC	
chr21	34635080	34635080	Heterozygous T		-	Novel	IFNAR2	INTRONIC	
chr21	34635080	34635080	Heterozygous T		-	Novel	IFNAR2	DOWNSTREAM	
chr21	34635080	34635080	Heterozygous T		-	Novel	IFNAR2	DOWNSTREAM	
chr21	34900917	34900918	Heterozygous -		A	Novel	GART	SPICE_SITE	
chr21	34900917	34900918	Heterozygous -		A	Novel	GART	SPICE_SITE	
chr21	34900917	34900918	Heterozygous -		A	Novel	GART	SPICE_SITE	
chr21	34900917	34900918	Heterozygous -		A	Novel	GART	SPICE_SITE	
chr21	34907637	34907637	Heterozygous A		-	Novel	GART	INTRONIC	
chr21	34907637	34907637	Heterozygous A		-	Novel	GART	INTRONIC	
chr21	34907637	34907637	Heterozygous A		-	Novel	GART	INTRONIC	
chr21	34907637	34907637	Heterozygous A		-	Novel	GART	INTRONIC	
chr21	34907637	34907637	Heterozygous A		-	Novel	GART	INTRONIC	
chr21	35757741	35757741	Heterozygous A		T	Novel	FAM185B	INTRONIC	
chr21	37413058	37413058	Heterozygous G		A	Novel	SETD4	INTRONIC	
chr21	37775127	37775127	Heterozygous C		T	Novel	CHAF1B	SYNONYMOUS_CODING	
chr21	38439597	38439597	Homozygous C		T	Novel	PIGP	NON_SYNONYMI>N	
chr21	38439597	38439597	Homozygous C		T	Novel	PIGP	NON_SYNONYMI>N	
chr21	38439597	38439597	Homozygous C		T	Novel	PIGP	EXONIC	
chr21	38536312	38536312	Heterozygous A		G	Novel	TTG3	INTRONIC	
chr21	38536312	38536312	Heterozygous A		G	Novel	TTG3	INTRONIC	
chr21	38877499	38877499	Homozygous A		G	Novel	DYRK1A	INTRONIC	
chr21	38877499	38877499	Homozygous A		G	Novel	DYRK1A	INTRONIC	
chr21	38877499	38877499	Homozygous A		G	Novel	DYRK1A	INTRONIC	
chr21	38877499	38877499	Homozygous A		G	Novel	DYRK1A	INTRONIC	
chr21	38877499	38877499	Homozygous A		G	Novel	DYRK1A	INTRONIC	
chr21	39772655	39772655	Heterozygous G		A	Novel	ERG	INTRONIC	
chr21	39772655	39772655	Heterozygous G		A	Novel	ERG	INTRONIC	
chr21	39772655	39772655	Heterozygous G		A	Novel	ERG	INTRONIC	
chr21	39772655	39772655	Heterozygous G		A	Novel	ERG	INTRONIC	
chr21	40646231	40646232	Heterozygous GA		-	Novel	BRWD1	INTRONIC	
chr21	40646231	40646232	Heterozygous GA		-	Novel	BRWD1	INTRONIC	
chr21	43221228	43221228	Heterozygous A		C	Novel	PRDM15	DOWNSTREAM	
chr21	43221228	43221228	Heterozygous A		C	Novel	PRDM15	3PRIME_UTR	
chr21	43277279	43277280	Heterozygous -		T	Novel	PRDM15	SPICE_SITE	
chr21	43277279	43277280	Heterozygous -		T	Novel	PRDM15	SPICE_SITE	
chr21	43279316	43279316	Heterozygous G		A	Novel	PRDM15	SYNONYMOUS_CODING	
chr21	43279316	43279316	Heterozygous G		A	Novel	PRDM15	INTRONIC	
chr21	43557778	43557778	Homozygous C		T	Novel	UMODL1	INTRONIC	
chr21	43557778	43557778	Homozygous C		T	Novel	UMODL1	INTRONIC	
chr21	43557778	43557778	Homozygous C		T	Novel	UMODL1	INTRONIC	
chr21	43708431	43708432	Heterozygous -		C	Novel	ABCG1	INTRONIC	
chr21	43708431	43708432	Heterozygous -		C	Novel	ABCG1	INTRONIC	
chr21	43708431	43708432	Heterozygous -		C	Novel	ABCG1	INTRONIC	
chr21	43708431	43708432	Heterozygous -		C	Novel	ABCG1	INTRONIC	
chr21	43708431	43708432	Heterozygous -		C	Novel	ABCG1	INTRONIC	
chr21	43708431	43708432	Heterozygous -		C	Novel	ABCG1	INTRONIC	
chr21	45709479	45709479	Heterozygous C		T	Novel	AIRE	INTRONIC	
chr21	45709479	45709479	Heterozygous C		T	Novel	AIRE	UPSTREAM	
chr21	45736185	45736185	Heterozygous G		A	Novel	PFKL	EXONIC	
chr21	45736185	45736185	Heterozygous G		A	Novel	PFKL	NON_SYNONYMI>H	
chr21	45959280	45959280	Heterozygous A		G	Novel	C21orf29	INTRONIC	
chr21	45959280	45959280	Heterozygous A		G	Novel	KRTAP10-1	NON_SYNONYMI>R	
chr21	46066466	46066466	Heterozygous T		C	Novel	C21orf29	INTRONIC	
chr21	46066466	46066466	Heterozygous T		C	Novel	KRTAP10-11	NON_SYNONYMI>R	
chr21	46716200	46716200	Heterozygous G		T	Novel	LOC642852	EXONIC	
chr21	47545369	47545370	Heterozygous -		C	Novel	COL6A2	INTRONIC	
chr21	47545369	47545370	Heterozygous -		C	Novel	COL6A2	INTRONIC	
chr21	47545369	47545370	Heterozygous -		C	Novel	COL6A2	INTRONIC	
chr22	16267115	16267115	Heterozygous T		C	Novel	POTEH	INTRONIC	
chr22	16287156	16287156	Heterozygous T		C	Novel	POTEH	INTRONIC	
chr22	16287372	16287372	Heterozygous C		T	Novel	POTEH	NON_SYNONYMI>I	

Chromosome	Start	End	Zygoty	Reference	SNP Call	dSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chr22	16287513	16287513	Heterozygous G		T	Novel	POTEH	NON_SYNONYMI>N	
chr22	16414031	16414031	Homozygous G		A	Novel		GENIC	
chr22	16871346	16871346	Heterozygous C		T	Novel		GENIC	
chr22	17447138	17447138	Heterozygous C		G	Novel	GAB4	NON_SYNONYMI>H	
chr22	18072807	18072807	Heterozygous G		A	Novel	SLC25A18	INTRONIC	
chr22	18910451	18910451	Heterozygous A		-	Novel	PRODH	SPICE_SITE	
chr22	18910451	18910451	Heterozygous A		-	Novel	PRODH	SPICE_SITE	
chr22	20132980	20132980	Heterozygous C		-	Novel		GENIC	
chr22	20761192	20761193	Heterozygous GC		TT	Novel	ZNF74	NON_SYNONYMI>N	
chr22	20761192	20761193	Heterozygous GC		TT	Novel	ZNF74	EXONIC	
chr22	21363262	21363262	Heterozygous C		T	Novel	FLJ39582	EXONIC	
chr22	21363262	21363262	Heterozygous C		T	Novel	MGC16703	EXONIC	
chr22	21407652	21407652	Heterozygous A		G	Novel	LOC400891	EXONIC	
chr22	21537977	21537977	Heterozygous G		A	Novel		GENIC	
chr22	21537999	21537999	Heterozygous A		G	Novel		GENIC	
chr22	21538046	21538046	Heterozygous T		C	Novel		GENIC	
chr22	21663593	21663593	Heterozygous G		C	Novel		GENIC	
chr22	22317166	22317166	Heterozygous A		G	Novel	TOP3B	NON_SYNONYMI>T	
chr22	22326235	22326235	Heterozygous A		C	Novel	TOP3B	INTRONIC	
chr22	22453425	22453425	Heterozygous C		G	Novel		GENIC	
chr22	22453469	22453469	Heterozygous C		T	Novel		GENIC	
chr22	22516766	22516767	Heterozygous -		TC	Novel		GENIC	
chr22	22681962	22681962	Heterozygous T		C	Novel		GENIC	
chr22	23114879	23114879	Heterozygous G		A	Novel		GENIC	
chr22	23247117	23247117	Heterozygous G		C	Novel		GENIC	
chr22	23282089	23282089	Heterozygous G		T	Novel		GENIC	
chr22	23282118	23282118	Heterozygous C		A	Novel		GENIC	
chr22	23285027	23285027	Heterozygous G		A	Novel		GENIC	
chr22	23541310	23541310	Heterozygous A		-	Novel	BCR	INTRONIC	
chr22	23541310	23541310	Heterozygous A		-	Novel	BCR	INTRONIC	
chr22	23541310	23541310	Heterozygous A		-	Novel	BCR	INTRONIC	
chr22	23541310	23541310	Heterozygous A		-	Novel	BCR	INTRONIC	
chr22	23556703	23556703	Heterozygous C		T	Novel	BCR	INTRONIC	
chr22	23556703	23556703	Heterozygous C		T	Novel	BCR	INTRONIC	
chr22	23742049	23742049	Heterozygous G		A	Novel	ZDHHC8P1	EXONIC	
chr22	24807730	24807730	Heterozygous C		T	Novel	SPECC1L	INTRONIC	
chr22	24807730	24807730	Heterozygous C		T	Novel	SPECC1L	INTRONIC	
chr22	24807730	24807730	Heterozygous C		T	Novel	SPECC1L	INTRONIC	
chr22	24940080	24940080	Heterozygous G		A	Novel	PWWL3	INTRONIC	
chr22	25153812	25153812	Heterozygous A		-	Novel	MYO18B	INTRONIC	
chr22	28219400	28219400	Heterozygous G		T	Novel	CCDC117	INTRONIC	
chr22	29169681	29169681	Heterozygous C		G	Novel	XBP1	INTRONIC	
chr22	29196043	29196044	Heterozygous -		T	Novel	XBP1	INTRONIC	
chr22	29196043	29196044	Heterozygous -		T	Novel	XBP1	INTRONIC	
chr22	30374873	30374873	Heterozygous T		-	Novel	MTMR3	INTRONIC	
chr22	30374873	30374873	Heterozygous T		-	Novel	MTMR3	INTRONIC	
chr22	30374873	30374873	Heterozygous T		-	Novel	MTMR3	INTRONIC	
chr22	30805444	30805444	Heterozygous C		T	Novel	SEC14L2	SYNONYMOUS_CODING	
chr22	30805444	30805444	Heterozygous C		T	Novel	SEC14L2	SYNONYMOUS_CODING	
chr22	31013374	31013374	Heterozygous C		T	Novel	TCN2	NON_SYNONYMI>M	
chr22	31013374	31013374	Heterozygous C		T	Novel	TCN2	NON_SYNONYMI>M	
chr22	31042716	31042716	Heterozygous G		A	Novel	SLC35E4	NON_SYNONYMI>N	
chr22	31942829	31942830	Heterozygous -		T	Novel	SFI1	INTRONIC	
chr22	31942829	31942830	Heterozygous -		T	Novel	SFI1	INTRONIC	
chr22	32081438	32081438	Heterozygous A		-	Novel	C22orf30	3PRIME_UTR	
chr22	32113164	32113164	Heterozygous T		G	Novel	C22orf30	NON_SYNONYMI>R	
chr22	32435506	32435507	Heterozygous AA		GC	Novel		GENIC	
chr22	32754513	32754513	Heterozygous G		-	Novel	RFPL3	INTRONIC	
chr22	32754513	32754513	Heterozygous G		-	Novel	RFPL3	INTRONIC	
chr22	32754513	32754513	Heterozygous G		-	Novel	RFPL3S	DOWNSTREAM	
chr22	32754513	32754513	Heterozygous G		-	Novel	RFPL3S	DOWNSTREAM	
chr22	33327428	33327428	Heterozygous G		A	Novel	SYN3		

Chromosome	Start	End	Zygoty	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chrX	38189777	38189778	Homozygous	-	A	Novel	RFRG	INTRONIC	
chrX	38189777	38189778	Homozygous	-	A	Novel	RFRG	INTRONIC	
chrX	38664580	38664580	Heterozygous	G	C	Novel	MID1IP1	SYNONYMOUS_CODING	
chrX	38664580	38664580	Heterozygous	G	C	Novel	MID1IP1	SYNONYMOUS_CODING	
chrX	38664580	38664580	Heterozygous	G	C	Novel	MID1IP1	SYNONYMOUS_CODING	
chrX	41394233	41394233	Heterozygous	C	T	Novel	CASK	SPLICE_SITE	
chrX	41394233	41394233	Heterozygous	C	T	Novel	CASK	INTRONIC	
chrX	41394233	41394233	Heterozygous	C	T	Novel	CASK	INTRONIC	
chrX	44386445	44386445	Heterozygous	A	-	Novel	FUNDC1	INTRONIC	
chrX	47041172	47041172	Heterozygous	G	A	Novel	RRM10	NON_SYNONYMI-V->M	
chrX	47041172	47041172	Heterozygous	G	A	Novel	RRM10	NON_SYNONYMI-V->M	
chrX	47088863	47088863	Heterozygous	A	G	Novel	UBA1	INTRONIC	
chrX	47088863	47088863	Heterozygous	A	G	Novel	UBA1	INTRONIC	
chrX	47101687	47101687	Heterozygous	G	A	Novel	USP11	SYNONYMOUS_CODING	
chrX	48053693	48053693	Heterozygous	G	T	Novel	SSX5	INTRONIC	
chrX	48053693	48053693	Heterozygous	G	T	Novel	SSX5	INTRONIC	
chrX	48270312	48270312	Heterozygous	A	-	Novel	SSX4B	SPLICE_SITE	
chrX	48270312	48270312	Heterozygous	A	-	Novel	SSX4B	SPLICE_SITE	
chrX	48674614	48674614	Heterozygous	C	T	Novel	HDAC6	SYNONYMOUS_CODING	
chrX	48973401	48973401	Heterozygous	T	C	Novel		GENIC	
chrX	49021412	49021412	Heterozygous	G	A	Novel	MAGIX	NON_SYNONYMI-R->H	
chrX	49021412	49021412	Heterozygous	G	A	Novel	MAGIX	INTRONIC	
chrX	49021412	49021412	Heterozygous	G	A	Novel	MAGIX	INTRONIC	
chrX	49021412	49021412	Heterozygous	G	A	Novel	MAGIX	NON_SYNONYMI-R->H	
chrX	52873752	52873752	Heterozygous	G	T	Novel	SSX7	INTRONIC	
chrX	52891820	52891820	Heterozygous	T	-	Novel	XAGE3	3PRIME_UTR	
chrX	52891820	52891820	Heterozygous	T	-	Novel	XAGE3	3PRIME_UTR	
chrX	52891732	52891732	Heterozygous	G	A	Novel	XAGE3	INTRONIC	
chrX	52891732	52891732	Heterozygous	G	A	Novel	XAGE3	INTRONIC	
chrX	53270950	53270950	Heterozygous	C	T	Novel	IQSEC2	INTRONIC	
chrX	53270950	53270950	Heterozygous	C	T	Novel	IQSEC2	INTRONIC	
chrX	53643886	53643886	Heterozygous	A	C	Novel	HUWE1	INTRONIC	
chrX	53652814	53652814	Heterozygous	A	C	Novel	HUWE1	INTRONIC	
chrX	56295439	56295440	Heterozygous	AC	-	Novel		GENIC	
chrX	76845449	76845449	Heterozygous	C	A	Novel	ATRX	INTRONIC	
chrX	76845449	76845449	Heterozygous	C	A	Novel	ATRX	INTRONIC	
chrX	77109494	77109494	Heterozygous	A	G	Novel	MAQT1	INTRONIC	
chrX	77245508	77245508	Heterozygous	T	C	Novel	ATP7A	INTRONIC	
chrX	77276413	77276413	Heterozygous	C	A	Novel	ATP7A	INTRONIC	
chrX	79283631	79283631	Heterozygous	C	-	Novel	TBX22	INTRONIC	
chrX	79283631	79283631	Heterozygous	C	-	Novel	TBX22	INTRONIC	
chrX	79283631	79283631	Heterozygous	C	-	Novel	TBX22	INTRONIC	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	UPSTREAM	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	UPSTREAM	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	UPSTREAM	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	5PRIME_UTR	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	5PRIME_UTR	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	UPSTREAM	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	INTRONIC	
chrX	91090441	91090443	Heterozygous	CTC	----	Novel	PCDH11X	5PRIME_UTR	
chrX	92965293	92965293	Heterozygous	A	G	Novel	FAM133A	3PRIME_UTR	
chrX	92965293	92965293	Heterozygous	A	G	Novel	FAM133A	3PRIME_UTR	
chrX	92965293	92965293	Heterozygous	A	G	Novel	FAM133A	3PRIME_UTR	
chrX	92965293	92965293	Heterozygous	A	G	Novel	FAM133A	3PRIME_UTR	
chrX	96171417	96171417	Heterozygous	T	-	Novel	DIAPH2	INTRONIC	
chrX	96171417	96171417	Heterozygous	T	-	Novel	DIAPH2	INTRONIC	
chrX	98551322	98551322	Heterozygous	T	G	Novel	PCDH19	NON_SYNONYMI-N->H	
chrX	98551322	98551322	Heterozygous	T	G	Novel	PCDH19	NON_SYNONYMI-N->H	
chrX	98551322	98551322	Heterozygous	T	G	Novel	PCDH19	NON_SYNONYMI-N->H	
chrX	100357456	100357456	Heterozygous	T	C	Novel	CEPFI	INTRONIC	
chrX	100530313	100530313	Heterozygous	A	-	Novel	TA7F7L	INTRONIC	
chrX	100530313	100530313	Heterozygous	A	-	Novel	TA7F7L	INTRONIC	
chrX	100668908	100668908	Heterozygous	T	-	Novel	HNRNP2	INTRONIC	
chrX	100668908	100668908	Heterozygous	T	-	Novel	HNRNP2	INTRONIC	
chrX	105139356	105139357	Heterozygous	----	AGAA	Novel	NRK	INTRONIC	
chrX	106069180	106069180	Heterozygous	G	T	Novel	TBC1D8B	INTRONIC	
chrX	106069180	106069180	Heterozygous	G	T	Novel	TBC1D8B	INTRONIC	
chrX	106108849	106108849	Heterozygous	A	G	Novel	TBC1D8B	NON_SYNONYMI-I->V	
chrX	106486286	106486287	Heterozygous	-	G	Novel	CXorf41	INTRONIC	
chrX	106486286	106486287	Heterozygous	-	G	Novel	CXorf41	INTRONIC	
chrX	107225417	107225417	Heterozygous	T	C	Novel	TEX13B	INTRONIC	
chrX	107400223	107400223	Heterozygous	T	G	Novel	COL4A6	3PRIME_UTR	
chrX	107400223	107400223	Heterozygous	T	G	Novel	COL4A6	3PRIME_UTR	
chrX	109416689	109416689	Heterozygous	T	-	Novel	TMEM164	3PRIME_UTR	
chrX	109416689	109416689	Heterozygous	T	-	Novel	TMEM164	3PRIME_UTR	
chrX	114141469	114141471	Heterozygous	AAG	----	Novel	HTR2C	NON_SYNONYMOUS_CODING	
chrX	114238593	114238593	Heterozygous	A	G	Novel	IL13RA2	3PRIME_UTR	

Chromosome	Start	End	Zygoty	Reference	SNP Call	dbSNP Match Category	Gene Symbol	Consequence	Amino Acid Change
chrX	118221283	118221283	Heterozygous	G	A	Novel	KIAA1210	NON_SYNONYMI-P->S	
chrX	118604553	118604554	Heterozygous	-	A	Novel	SLC25A5	INTRONIC	
chrX	118679316	118679316	Heterozygous	A	-	Novel		GENIC	
chrX	119678897	119678897	Heterozygous	A	-	Novel	CUL4B	INTRONIC	
chrX	119678897	119678897	Heterozygous	A	-	Novel	CUL4B	INTRONIC	
chrX	122756727	122756727	Heterozygous	G	A	Novel	THOC2	INTRONIC	
chrX	123515067	123515067	Heterozygous	G	C	Novel	ODZ1	SYNONYMOUS_CODING	
chrX	123515067	123515067	Heterozygous	G	C	Novel	ODZ1	SYNONYMOUS_CODING	
chrX	123515067	123515067	Heterozygous	G	C	Novel	ODZ1	SYNONYMOUS_CODING	
chrX	12857376	12857376	Heterozygous	C	T	Novel	SMARCA1	SPRIME_UTR	
chrX	12857376	12857376	Heterozygous	C	T	Novel	SMARCA1	SPRIME_UTR	
chrX	12879144	12879144	Heterozygous	G	A	Novel	XPNPEP2	INTRONIC	
chrX	129265512	129265512	Heterozygous	T	C	Novel	AIFM1	INTRONIC	
chrX	129265512	129265512	Heterozygous	T	C	Novel	AIFM1	INTRONIC	
chrX	129265512	129265512	Heterozygous	T	C	Novel	AIFM1	INTRONIC	
chrX	129265512	129265512	Heterozygous	T	C	Novel	AIFM1	INTRONIC	
chrX	129265512	129265512	Heterozygous	T	C	Novel	AIFM1	INTRONIC	
chrX	130420386	130420386	Heterozygous	G	A	Novel	IGSF1	INTRONIC	
chrX	130420386	130420386	Heterozygous	G	A	Novel	IGSF1	INTRONIC	
chrX	130420386	130420386	Heterozygous	G	A	Novel	IGSF1	INTRONIC	
chrX	130420386	130420386	Heterozygous	G	A	Novel	IGSF1	INTRONIC	
chrX	131216421	131216421	Heterozygous	A	G	Novel	FRMD7	NON_SYNONYMI-L->P	
chrX	134025422	134025422	Heterozygous	A	-	Novel	MOSPD1	INTRONIC	
chrX	135029298	135029298	Heterozygous	C	T	Novel		GENIC	
chrX	135574074	135574074	Heterozygous	T	G	Novel	BRS3	INTRONIC	
chrX	138708211	138708211	Heterozygous	G	A	Novel	MCF2	INTRONIC	
chrX	138708211	138708211	Heterozygous	G	A	Novel	MCF2	INTRONIC	
chrX	138708211	138708211	Heterozygous	G	A	Novel	MCF2	INTRONIC	
chrX	138708211	138708211	Heterozygous	G	A	Novel	MCF2	INTRONIC	
chrX	138708211	138708211	Heterozygous	G	A	Novel	MCF2	INTRONIC	
chrX	138880934	138880934	Heterozygous	C	A	Novel	ATP11C	INTRONIC	
chrX	138880934	138880934	Heterozygous	C	A	Novel	ATP11C	INTRONIC	
chrX	138882173	138882174	Heterozygous	CC	-C	Novel	ATP11C	INTRONIC	
chrX	138882173	138882174	Heterozygous	CC	-	Novel	ATP11C	INTRONIC	
chrX	138882173	138882174	Heterozygous	CC	-C	Novel	ATP11C	INTRONIC	
chrX	140993801	140993801	Heterozygous	G	C	Novel	MAGEG1	NON_SYNONYMI-R->P	
chrX	140993827	140993827	Heterozygous	A	C	Novel	MAGEG1	NON_SYNONYMI-I->L	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	148048208	148048210	Heterozygous	TTT	----	Novel	AF2	INTRONIC	
chrX	151870296	151870296	Heterozygous	T	C	Novel	MAGEA6	3PRIME_UTR	
chrX	151870296	151870296	Heterozygous	T	C	Novel	MAGEA6	3PRIME_UTR	
chrX	152087447	152087447	Heterozygous	G	A	Novel	ZNF185	INTRONIC	
chrX	152087447	152087447	Heterozygous	G	A	Novel	ZNF185	INTRONIC	
chrX	152087447	152087447	Heterozygous	G	A	Novel	ZNF185	INTRONIC	
chrX	152087447	152087447	Heterozygous	G	A	Novel	ZNF185	INTRONIC	
chrX	152087447	152087447	Heterozygous	G	A	Novel	ZNF185	INTRONIC	
chrX	152087447	152087447	Heterozygous	G	A	Novel	ZNF185	INTRONIC	
chrX	153216187	153216187	Heterozygous	C	T	Novel	HCF1	INTRONIC	
chrX	153592315	153592315	Heterozygous	C	-	Novel	FLNA	INTRONIC	
chrX	153592315	153592315	Heterozygous	C	-	Novel	FLNA	INTRONIC	

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分担研究報告書

反復胞状奇胎の分子遺伝学的解析

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研究要旨

全胞状奇胎は、雄核発生（父ゲノムのみを有する二倍体）としての特徴を有しているのに対し、反復胞状奇胎（反復胞状奇胎）は、一見正常な二倍体であるが、DNAメチル化の異常を伴い、*NALP7*遺伝子変異と関連している事が明らかとなった。従来の形態学的診断では、反復胞状奇胎を通常の全胞状奇胎と区別する事は不可能であり、反復胞状奇胎の診断には、分子遺伝学的な解析が有効かつ必須である。しかし本邦の反復胞状奇胎症例は、疫学的報告は散見されるが、分子遺伝学的診断法について系統的に検討解析した研究は見当たらない。

そこで本研究計画では、候補症例検体および臨床情報を収集し、母 *NALP7* 遺伝子の変異解析を行い、反復胞状奇胎の発症起源を分子遺伝学的に確定する。これこれらの解析結果を統合し、本邦の反復胞状奇胎症例に最適な確定診断法の開発と、臨床的特徴を抽出する事を目的とする。試験的に行った習慣流産症例の解析では、5つの新規遺伝子多型が得られた。また本年度の研究で、本邦初の *NALP7* 遺伝子ホモ変異症例の同定に成功した。

本研究の進捗により、発症起源に遡って胞状奇胎を正確に分類・確定診断することが可能となる。最近新たな候補因子遺伝子 (*c6orf221* 遺伝子) も報告されており、*NALP7* 遺伝子変異と併せ、反復胞状奇胎周辺類縁疾患との関連解明も期待される。

A. 研究目的

初年度は、日本人集団で頻度が高いと考えられる、公的なデータベースに登録されていない候補因子遺伝子多型を同定したが、既知の候補因子遺伝子変異を同定することはできなかった。そこで本年度は、既知変異以外の配列の特徴の解析を中心に行った。

本邦では、全胞状奇胎（全胞状奇胎）に続く妊娠が再び全胞状奇胎となる確率は、通常的全胞状奇胎発生頻度の10倍前後で、反復率は2-5%に上るとされ

ている。近年の胞状奇胎の発症数は、出生数1,000人に対し1例前後であり、年間の出生数は100万人前後である事から、毎年数十人の患者が、胞状奇胎を反復して発症していると推定される。

最近の遺伝学的研究から、反復胞状奇胎（反復胞状奇胎）は、通常的全胞状奇胎と極めて異なる病因を有することが明らかとなった。2006年に、家族性反復胞状奇胎の解析から、母親の *NALP7* 遺伝子変異が反復胞状奇胎の原因であることが同定された (*Nat Genet.* 2006;38:300-2)。さらに最新の報告で

は、孤発症例であっても、2回以上の奇胎を繰り返す症例のほとんどに、*NALP7* 遺伝子に変異が存在することが示された。(Hum Mutat. 2009;30:E629-39, Hum Mol Genet. 2009;18:888-97) その他に、反復胞状奇胎は両親ゲノムを有する一見正常な核型で(通常の全胞状奇胎は父ゲノムのみを有する)、DNAメチル化異常を伴う等の特徴も明らかになった。これらの分子遺伝学的診断と従来の形態的診断を併用すれば、反復胞状奇胎の確定診断が可能となる。しかし本邦では、反復胞状奇胎症例の疫学的報告は散見されるが、病因病態から管理法までを系統的に検討解析した研究は見当たらない。

そこで本研究計画では、候補患者検体および病歴等の臨床情報を収集する。また、*NALP7* 遺伝子をはじめとする遺伝子変異解析を行い、併せて、反復胞状奇胎組織の発症起源を分子遺伝学的に確定する。これらの解析結果を統合し、本邦の反復胞状奇胎症例に最適な確定診断法の開発と、臨床的特徴を抽出する事を目的とする。

B. 研究方法

本邦での反復胞状奇胎症例の実態は把握されておらず、分子診断法の有効性も検証されていない。そこで、分担研究者らの協力を得て、1) 患者検体および病歴等の臨床情報を収集する。また、2) 母親末梢血の *NALP7* 遺伝子の変異解析を行い、3) 反復胞状奇胎組織の分子遺伝学発生起源診断を行う。

上記目的を達成するために、次頁に記した以下の計画に沿って研究を推進す

る。

1) 検体と臨床情報の収集

症例照会により反復胞状奇胎候補症例が見出されたら、患者の末梢血を抗凝固剤入りの採血管で5ml採取し、末梢血リンパ球からゲノムDNAを回収する。奇胎組織は、固定標本が保存されていれば、標本からゲノムDNAを回収する。検体収集と併せて、詳細な臨床情報(特に妊娠分娩歴、家族歴、治療経過)を収集する。すべての情報は、連結可能匿名化を行い、個人情報厳密かつ適正に扱う。

研究代表者秦は現在、類似の遺伝子解析研究を行っており、ゲノムDNA回収技術及び個人情報等の管理体制はすでに確立している。

2) *NALP7* 遺伝子変異解析

海外の孤発症例および家族性症例で同定された母体の *NALP7* 遺伝子変異の有無を、サンガー法による遺伝子配列解析装置を用いて解析する。海外の症例で同定された遺伝子変異は多岐にわたっており(2009年7月時点で約30種類)、日本人集団にもおそらく日本人集団で頻度の高い新規の多型が、初年度の解析で多数見つかった。これらの知見を元に、症例で特異的な候補因子遺伝子配列の特徴を同定する。

3) 胞状奇胎組織の分子遺伝学的解析

3.1) 多型マーカー解析による発症起源の決定

胞状奇胎組織のゲノムDNAと、母末梢血の多型マーカー解析を行い、その比較により、奇胎の発症起源(雄核発生、正常2倍体、2精子受精3倍体、等)の

決定を行う。

3.2) DNA メチル化解析

海外の反復胞状奇胎組織はいずれも、インプリンティング遺伝子の DNA メチル化異常を伴う。研究代表者らは、すでにヒトインプリンティング遺伝子の制御領域を含む 32 箇所 DNA メチル化状態を網羅的に解析する系を確立している。この解析系を用い、反復胞状奇胎絨毛組織の網羅的 DNA メチル化解析を行う。

C. 研究結果

結果 1) 候補遺伝子変異診断法の開発

初年度は、*NALP7* 遺伝子約 30,000 塩基対の中から、海外で報告のあった 10 領域で、合計 5,796 塩基の配列解析を行った。その結果、合計 8 箇所、未知の、しかしおそらくは日本人集団では比較的高頻度に同定される *NALP7* 遺伝子多型を見出した。その一方で、症例特異的な異常を乱すことができなかった。本年はさらに解析範囲を広げ、*NALP7* 遺伝子にホモ変異を同定した(資料 1)。

結果 2) 胞状奇胎組織の分子遺伝学的解析

上記の、*NALP7* 遺伝子ホモ変異を同定した症例の絨毛組織は、海外の類似症例結果から、見かけ上は父ゲノムと母ゲノムを有する正常二倍体であることが予想される。しかし、絨毛組織標本から回収されたゲノム DNA を用いたマーカー解析では、父由来の遺伝マーカーを検出できず、確定診断に至らなかった。回収されたゲノム DNA のほとんどが母親由来(脱落膜組織由来)で

あったと考えられる。

結果 3) 胞状奇胎組織の DNA メチル化診断

我々が独自に確立した、インプリンティング遺伝子関連ゲノム領域の DNA メチル化スクリーニング法を用い、収集した反復胞状奇胎組織および「のう胞化絨毛組織を呈するが両親由来のゲノムを有する正常二倍体」の絨毛ゲノム DNA の解析を行ったが、現在までのところ、これらの症例の DNA メチル化状態は、正常絨毛(人工妊娠中絶症例)の DNA メチル化状態と明らかな差異は認められなかった。

D. 考察

最終年度である本年度の研究により、海外症例で関連が報告されている *NALP7* 遺伝子にホモ変異を有する症例を同定することに成功した。本邦では、反復胞状奇胎症例を含め、奇胎症例の遺伝子診断は行われていない。本研究に因り、本邦で初めて、反復胞状奇胎の関連候補因子に、新規のホモ変異を見出すことに成功した。また、本研究により、海外では報告されていない未知の遺伝子多型(変異)を複数見出している。これらの変異は、症例に特異的な異常か、あるいは一定の頻度で正常集団にも見出される多型なのかは、今後のさらなる解析が必要であるが、いずれにせよ、日本人集団に特有の遺伝子型と考えられ、本邦症例に最適化した診断法の確立に必須の貴重な知見が得られたと考える。こ

海外の報告では、家族性反復胞状奇胎症例のみならず、家族歴を伴わない孤

発例・正常な妊娠歴のある症例・奇胎と診断されていなかった不育症症例などでも、*NALP7* 遺伝子変異が同定されていることから、本研究は、習慣流産など反復胎状奇胎以外の異常妊娠解析にも展開できる可能性がある。

また、最近 *c6orf221* 遺伝子変異との関連も海外症例では報告されており、今後候補遺伝子を増やして解析が必要であろう。

現在、「染色体転座に起因する習慣流産」は着床前診断の審査対象とされているが、反復胎状奇胎は習慣流産よりもはるかに母体の重篤な合併症リスクが高く、医学的な見地からは着床前診断を考慮すべき疾患であると考えられる。本研究成果の展開応用により、微量組織（一細胞）を用いた反復胎状奇胎の分子遺伝学的診断法が可能であり、確実な着床前診断法の確立が期待できる。

現在、反復胎状奇胎の分子遺伝学的診断に成功しているのは海外の数グループのみで、国内で類似の検討は行われていない。従来奇胎は、形態学的分類に従って分類が成されてきたが、分子遺伝学的な病因病態による細分類・再分類を行えば、発症後の妊娠転帰や合併症発症率が異なる可能性が十分考えられる。本研究で得られる知見は、絨毛性疾患の診断管理法に、多大な貢献を成すことが期待される。海外の報告では、2回以上の全胎状奇胎を反復する症例の大部分に、*NALP7* 遺伝子に変異が存在することが示された。本邦の症例も、同様の分子遺伝学的解析手法により、確定診断を開発できると予想される。また、確定診断に基づいた新たな分類で臨床経過を再評価し、適正な反

復胎状奇胎症例の管理法が提唱できると考えられる。

E. 結論

分担研究者らの協力を得て、反復胎状奇胎疑い症例の、現在臨床情報と検体の収集およびその解析を進めている。

最終年度である本年度は、分子遺伝学的診断法を確立し、候補因子遺伝子である *NALP7* に既知の変異を認めない反復胎状奇胎候補症例を対象に、解析領域を拡大して遺伝子解析を行った。これらの解析から、新規のホモ変異（ナンセンス変異）の同定に成功した。昨年度までに同定した未知の *NALP7* 遺伝子多型、すなわち、日本人集団で比較的頻度の高いと推測される多型が8箇所と併せ、本邦症例を分子遺伝学的に解析するための体制は整ったと考える。

F. 健康危険情報

特記事項なし。

G. 研究発表

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なし

研究成果刊行に関する一覧表