Supplementary table 4a. Rare predicted loss of function variants found in cases

Chr = chromosome, Ref = reference allele, Alt = alternative allele, CADD = Combined Annotation Dependent Depletion score, Het/hom = heterozygous/homozygous, cDNA = complementary DNA, CDS = coding sequence, ExAC_AF = Exome Aggregation Consortium allele frequency, Seq_AF = Sequenced samples allele frequency, rsid = Reference SNP cluster ID

Chr	Position	Ref	Alt	Annotation	GeneSymbol	CADD	Het/hom alt allele	cDNA_position	CDS_position	AminoAcid_position	AminoAcid_change	CodonChange	ExonNumber	Gene	Transcript	ExAC_AF	Seq_AF	rsid
1	196643061	G	A	missense_variant	*CFH*	27.2	het	559/4127	319/3696	107/1231	G/S	Ggt/Agt	42816	3075	NM_000186.3	NA	1.04E-04	-
1	196643093	G	A	splice_donor_variant	*CFH*	20.10	het	-/4127	-/3696	-/1231	-	-	-	3075	NM_000186.3	NA	1.04E-04	-
1	196646789	A	ATT	frameshift_variant	*CFH*	NA	het	851-852/4127	611-612/3696	204/1231	K/NX	aag/aaTTg	41415	3075	NM_000186.3	NA	2.08E-04	-
1	196646789	A	ATT	frameshift_variant	*CFH*	NA	het	851-852/4127	611-612/3696	204/1231	K/NX	aag/aaTTg	41415	3075	NM_000186.3	NA	2.08E-04	-
1	196658617	C	G	stop_gained	*CFH*	15.08	het	1272/4127	1032/3696	344/1231	Y/*	taC/taG	41507	3075	NM_000186.3	NA	1.04E-04	-
1	196748330	CTATTTCA	C	frameshift_variant	*CFHR3*	NA	het	185-191/1457	98-104/810	33-35/269	LFH/X	cTATTTCAt/ct	41309	10878	NM_001166624.1	3.46E-05	3.12E-04	-
1	196748330	CTATTTCA	C	frameshift_variant	*CFHR3*	NA	het	185-191/1457	98-104/810	33-35/269	LFH/X	cTATTTCAt/ct	41309	10878	NM_001166624.1	3.46E-05	3.12E-04	-
1	196748929	A	T	stop_gained&splice_region_variant	*CFHR3*	11.55	het	343/1457	256/810	86/269	K/*	Aaa/Taa	41337	10878	NM_001166624.1	6.10E-05	2.08E-04	-
1	196883778	G	A	stop_gained	*CFHR4*	18.12	het	1471/2181	1334/1737	445/578	W/*	tGg/tAg	41495	10877	NM_001201550.2	1.55E-03	2.08E-04	rs376896638
1	196963306	G	GA	frameshift_variant	*CFHR5*	NA	het	655-656/2829	527-528/1710	176/569	R/RX	aga/agAa	41373	81494	NM_030787.3	2.47E-05	1.04E-04	-
1	197009779	C	T	missense_variant	*F13B*	21.5	het	1869/2217	1825/1986	609/661	G/S	Ggt/Agt	43051	2165	NM_001994.2	NA	1.04E-04	-
1	197036552	A	G	splice_donor_variant	*F13B*	6.958	het	-/2472	-/1746	-/581	-	-	-	2165	XM_005244955.1	NA	4.16E-04	-
1	197063273	T	C	missense_variant	*ASPM*	25.2	het	4527/6133	4270/5679	1424/1892	R/G	Aga/Gga	19/27	259266	NM_001206846.1	NA	1.04E-04	-
2	228109081	G	A	splice_donor_variant	*COL4A3*	17.43	het	-/8097	-/5013	-/1670	-	-	-	1285	NM_000091.4	8.28E-06	2.08E-04	rs202001097
2	228124001	T	G	splice_acceptor_variant&non_coding_transcript_variant	*LOC654841*	7.533	het	-/2502	-	-	-	-	-	654841	NR_102371.1	NA	1.04E-04	-
2	228124533	G	T	stop_gained	*COL4A3*	39	het	1216/8097	1054/5013	352/1670	E/*	Gaa/Taa	19/52	1285	NM_000091.4	NA	1.04E-04	-
2	228173618	C	T	missense_variant	*COL4A3*	20.3	het	4628/8097	4466/5013	1489/1670	T/I	aCt/aTt	49/52	1285	NM_000091.4	2.48E-05	2.08E-04	rs200818438
2	228176554	C	T	missense_variant	*COL4A3*	23.2	het	5143/8097	4981/5013	1661/1670	R/C	Cgc/Tgc	52/52	1285	NM_000091.4	3.06E-04	6.24E-04	rs201697532
3	64635449	C	T	missense_variant	*ADAMTS9*	30	het	1501/7313	1469/5808	490/1935	G/D	gGt/gAt	14885	56999	NM_182920.1	2.31E-04	8.32E-04	rs147650040
3	64635449	C	T	missense_variant	*ADAMTS9*	30	het	1501/7313	1469/5808	490/1935	G/D	gGt/gAt	14885	56999	NM_182920.1	2.31E-04	8.32E-04	rs147650040
3	64635449	C	T	missense_variant	*ADAMTS9*	30	het	1501/7313	1469/5808	490/1935	G/D	gGt/gAt	14885	56999	NM_182920.1	2.31E-04	8.32E-04	rs147650040
3	64635449	C	T	missense_variant	*ADAMTS9*	30	het	1501/7313	1469/5808	490/1935	G/D	gGt/gAt	14885	56999	NM_182920.1	2.31E-04	8.32E-04	rs147650040
3	64635449	C	T	missense_variant	*ADAMTS9*	30	het	1501/7313	1469/5808	490/1935	G/D	gGt/gAt	14885	56999	NM_182920.1	2.31E-04	8.32E-04	rs147650040
3	99509699	T	C	missense_variant	*COL8A1*	27	het	432/3091	173/2235	58/744	V/A	gTa/gCa	42830	1295	NM_001850.4	2.97E-04	2.08E-04	-
4	110605798	G	A	missense_variant	*CCDC109B*	20.10	het	951/1274	812/1011	271/336	R/Q	cGa/cAa	42894	55013	NM_017918.4	2.22E-04	5.20E-04	rs201083400
4	110610551	T	G	missense_variant	*CASP6*	32	het	895/1661	817/882	273/293	K/Q	Aag/Cag	42923	839	NM_001226.3	3.30E-05	2.08E-04	rs200347007
4	110615797	C	T	missense_variant	*CASP6*	21.2	het	445/1661	367/882	123/293	E/K	Gaa/Aaa	42862	839	NM_001226.3	9.06E-05	1.04E-04	-
4	110629624	C	A	start_lost	*CASP6*	5.527	het	191/1510	3/858	1/285	M/I	atG/atT	41280	839	XM_005263271.1	NA	1.04E-04	-
4	110635603	C	T	missense_variant	*PLA2G12A*	28.7	het	777/5235	500/570	167/189	G/D	gGt/gAt	42829	81579	NM_030821.4	8.24E-06	1.04E-04	-
4	110638764	A	G	missense_variant	*PLA2G12A*	20.8	het	668/5235	391/570	131/189	C/R	Tgc/Cgc	42798	81579	NM_030821.4	5.27E-04	9.36E-04	rs41278045
4	110638764	A	G	missense_variant	*PLA2G12A*	20.8	het	668/5235	391/570	131/189	C/R	Tgc/Cgc	42798	81579	NM_030821.4	5.27E-04	9.36E-04	rs41278045
4	110638764	A	G	missense_variant	*PLA2G12A*	20.8	het	668/5235	391/570	131/189	C/R	Tgc/Cgc	42798	81579	NM_030821.4	5.27E-04	9.36E-04	rs41278045
4	110638764	A	G	missense_variant	*PLA2G12A*	20.8	het	668/5235	391/570	131/189	C/R	Tgc/Cgc	42798	81579	NM_030821.4	5.27E-04	9.36E-04	rs41278045
4	110638764	A	G	missense_variant	*PLA2G12A*	20.8	het	668/5235	391/570	131/189	C/R	Tgc/Cgc	42798	81579	NM_030821.4	5.27E-04	9.36E-04	rs41278045
4	110650804	G	C	missense_variant	*PLA2G12A*	34	het	439/5235	162/570	54/189	D/E	gaC/gaG	42739	81579	NM_030821.4	3.30E-05	3.12E-04	rs376984776
4	110650804	G	C	missense_variant	*PLA2G12A*	34	het	439/5235	162/570	54/189	D/E	gaC/gaG	42739	81579	NM_030821.4	3.30E-05	3.12E-04	rs376984776
4	110662159	C	G	missense_variant	*CFI*	24	het	1850/2161	1642/1752	548/583	E/Q	Gaa/Caa	13/13	3426	NM_000204.3	7.17E-04	7.28E-04	rs7437875
4	110662159	C	G	missense_variant	*CFI*	24	het	1850/2161	1642/1752	548/583	E/Q	Gaa/Caa	13/13	3426	NM_000204.3	7.17E-04	7.28E-04	rs7437875
4	110667377	C	G	splice_donor_variant	*CFI*	14.05	het	-/2161	-/1752	-/583	-	-	-	3426	NM_000204.3	3.30E-05	2.08E-04	rs368555424
4	110667377	C	G	splice_donor_variant	*CFI*	14.05	het	-/2161	-/1752	-/583	-	-	-	3426	NM_000204.3	3.30E-05	2.08E-04	rs368555424
4	110670416	T	G	missense_variant	*CFI*	21.2	het	1314/2161	1106/1752	369/583	Y/S	tAt/tCt	43021	3426	NM_000204.3	8.24E-06	1.04E-04	-
4	110670478	C	T	splice_acceptor_variant	*CFI*	19.35	het	-/2161	-/1752	-/583	-	-	-	3426	NM_000204.3	NA	1.04E-04	-
4	110681679	C	T	missense_variant&splice_region_variant	*CFI*	25.5	het	980/2161	772/1752	258/583	A/T	Gca/Aca	42868	3426	NM_000204.3	1.07E-04	3.12E-04	rs199688124
4	110681679	C	T	missense_variant&splice_region_variant	*CFI*	25.5	het	980/2161	772/1752	258/583	A/T	Gca/Aca	42868	3426	NM_000204.3	1.07E-04	3.12E-04	rs199688124
4	110681687	C	T	missense_variant	*CFI*	20.7	het	972/2161	764/1752	255/583	C/Y	tGt/tAt	42868	3426	NM_000204.3	8.24E-06	1.04E-04	-
4	110681732	G	C	missense_variant	*CFI*	21.3	het	927/2161	719/1752	240/583	A/G	gCc/gGc	42868	3426	NM_000204.3	2.72E-04	4.16E-04	rs146444258
4	110681732	G	C	missense_variant	*CFI*	21.3	het	927/2161	719/1752	240/583	A/G	gCc/gGc	42868	3426	NM_000204.3	2.72E-04	4.16E-04	rs146444258
4	110681732	G	C	missense_variant	*CFI*	21.3	het	927/2161	719/1752	240/583	A/G	gCc/gGc	42868	3426	NM_000204.3	2.72E-04	4.16E-04	rs146444258
4	110739212	A	G	missense_variant	*GAR1*	24.7	het	642/1267	335/654	112/217	K/R	aAa/aGa	42801	54433	NM_018983.3	1.15E-04	4.16E-04	rs143266987
4	110739212	A	G	missense_variant	*GAR1*	24.7	het	642/1267	335/654	112/217	K/R	aAa/aGa	42801	54433	NM_018983.3	1.15E-04	4.16E-04	rs143266987
4	110756579	G	A	missense_variant	*RRH*	23.3	het	389/1558	355/1014	119/337	V/M	Gtg/Atg	42801	10692	NM_006583.2	2.72E-04	2.08E-04	rs200736180
4	110757186	T	TA	stop_gained&frameshift_variant	*RRH*	NA	het	455-456/1558	421-422/1014	141/337	Y/*	tac/tAac	41370	10692	NM_006583.2	NA	1.04E-04	-
5	39306855	G	C	stop_gained	*C9*	37	het	1369/2693	1280/1680	427/559	S/*	tCa/tGa	41527	735	NM_001737.3	8.24E-06	2.08E-04	rs121909594
5	39311320	T	C	missense_variant	*C9*	21.6	het	1119/2693	1030/1680	344/559	T/A	Act/Gct	42927	735	NM_001737.3	1.65E-05	1.04E-04	-
5	39388465	A	T	missense_variant	*DAB2*	26.4	het	1160/4601	629/2313	210/770	V/D	gTt/gAt	42993	1601	NM_001343.3	1.24E-04	4.16E-04	rs140161347
5	39388465	A	T	missense_variant	*DAB2*	26.4	het	1160/4601	629/2313	210/770	V/D	gTt/gAt	42993	1601	NM_001343.3	1.24E-04	4.16E-04	rs140161347
6	31743196	CTG	C	stop_gained&frameshift_variant	*VWA7*	NA	het	760-761/2914	522-523/2676	174-175/891	YS/*	taCAgt/tagt	41380	80737	NM_025258.2	NA	1.04E-04	-
6	31743869	C	G	missense_variant	*VWA7*	23.7	het	623/2914	385/2676	129/891	D/H	Gat/Cat	42811	80737	NM_025258.2	NA	1.04E-04	rs373092700
6	31747216	C	T	missense_variant	*VARS*	21.2	het	3809/4294	3386/3795	1129/1264	R/Q	cGg/cAg	28/30	7407	NM_006295.2	2.97E-04	5.20E-04	rs150145769
6	31748836	A	C	missense_variant	*VARS*	22.6	het	3032/4294	2609/3795	870/1264	V/G	gTc/gGc	22/30	7407	NM_006295.2	NA	1.04E-04	-
6	31760654	G	A	missense_variant	*VARS*	22.6	het	964/4294	541/3795	181/1264	R/C	Cgc/Tgc	42855	7407	NM_006295.2	6.90E-03	2.08E-04	rs35196751
6	31762948	A	C	missense_variant	*VARS*	25.9	het	470/4294	47/3795	16/1264	L/R	cTc/cGc	10990	7407	NM_006295.2	2.66E-04	2.08E-04	rs200831491
6	31762948	A	C	missense_variant	*VARS*	25.9	het	470/4294	47/3795	16/1264	L/R	cTc/cGc	10990	7407	NM_006295.2	2.66E-04	2.08E-04	rs200831491
6	31778258	C	T	missense_variant	*HSPA1L*	22.3	het	1675/2537	1492/1926	498/641	G/S	Ggc/Agc	42768	3305	NM_005527.3	8.24E-06	1.04E-04	-
6	31807421	GA	G	frameshift_variant	*C6orf48*	NA	het	723/842	210/228	70/75	G/X	ggA/gg	41398	50854	NM_001040437.1	NA	3.12E-04	-
6	31829005	C	T	missense_variant	*NEU1*	28.3	het	731/2071	575/1248	192/415	G/D	gGc/gAc	42800	4758	NM_000434.3	NA	2.08E-04	-
6	31829005	C	T	missense_variant	*NEU1*	28.3	het	731/2071	575/1248	192/415	G/D	gGc/gAc	42800	4758	NM_000434.3	NA	2.08E-04	-
6	31829865	C	G	missense_variant	*NEU1*	35	het	419/2071	263/1248	88/415	G/A	gGc/gCc	42772	4758	NM_000434.3	6.47E-03	2.08E-04	rs34712643
6	31833107	A	C	missense_variant	*SLC44A4*	20.2	het	1685/2508	1619/2007	540/668	M/R	aTg/aGg	16/20	80736	NM_001178044.1	NA	1.04E-04	rs150931872
6	31864215	CCT	C	frameshift_variant	*EHMT2*	NA	het	415-416/3968	405-406/3633	135-136/1210	TG/TX	acAGgg/acgg	41391	10919	NM_006709.3	NA	1.04E-04	-
6	31911055	G	A	missense_variant	*C2*	27.8	het	1678/2838	1414/2259	472/752	A/T	Gcc/Acc	43057	717	NM_000063.4	4.49E-04	1.04E-04	rs142243595
6	31911920	C	G	missense_variant	*C2*	26.9	het	2083/2838	1819/2259	607/752	L/V	Ctg/Gtg	15/18	717	NM_000063.4	8.47E-06	1.04E-04	-
6	31914933	C	T	stop_gained	*CFB*	37	het	726/2627	448/2295	150/764	R/*	Cga/Tga	41350	629	NM_001710.5	NA	1.04E-04	-
6	31929831	G	A	missense_variant&splice_region_variant	*SKIV2L*	37	het	1453/4170	1064/3741	355/1246	R/H	cGc/cAc	43036	6499	NM_006929.4	1.69E-05	1.04E-04	-
6	31934780	G	C	splice_acceptor_variant	*SKIV2L*	14.77	het	-/4170	-/3741	-/1246	-	-	-	6499	NM_006929.4	NA	1.04E-04	-
6	31934896	C	CA	frameshift_variant	*SKIV2L*	NA	het	2845-2846/4170	2456-2457/3741	819/1246	T/TX	aca/acAa	20/28	6499	NM_006929.4	NA	3.12E-04	-
6	31934896	C	CA	frameshift_variant	*SKIV2L*	NA	het	2845-2846/4170	2456-2457/3741	819/1246	T/TX	aca/acAa	20/28	6499	NM_006929.4	NA	3.12E-04	-

6	31948275	G	A	stop_gained	*STK19*	39	het	979/1620	851/1095	284/364	W/*	tGg/tAg	41432	8859	NM_004197.1	1.26E-04	2.08E-04	rs140503088
6	32012816	G	A	missense_variant	*TNXB*	20.8	het	11090/13125	10888/12729	3630/4242	R/C	Cgc/Tgc	32/44	7148	NM_019105.6	4.27E-05	1.04E-04	-
6	32021216	C	T	missense_variant	*TNXB*	23.9	het	8936/13125	8734/12729	2912/4242	G/S	Ggc/Agc	25/44	7148	NM_019105.6	6.72E-04	9.36E-04	rs200708257
6	32035615	C	T	missense_variant	*TNXB*	20.5	het	6569/13125	6367/12729	2123/4242	D/N	Gac/Aac	18/44	7148	NM_019105.6	5.55E-04	5.20E-04	rs201764050
6	32035615	C	T	missense_variant	*TNXB*	20.5	het	6569/13125	6367/12729	2123/4242	D/N	Gac/Aac	18/44	7148	NM_019105.6	5.55E-04	5.20E-04	rs201764050
6	32035615	C	T	missense_variant	*TNXB*	20.5	het	6569/13125	6367/12729	2123/4242	D/N	Gac/Aac	18/44	7148	NM_019105.6	5.55E-04	5.20E-04	rs201764050
6	32038079	G	T	missense_variant	*TNXB*	20	het	5305/13125	5103/12729	1701/4242	F/L	ttC/ttA	14/44	7148	NM_019105.6	7.03E-04	2.08E-04	rs201360247
6	32057054	G	A	stop_gained	*TNXB*	43	het	2663/13125	2461/12729	821/4242	R/*	Cga/Tga	14731	7148	NM_019105.6	2.48E-05	2.08E-04	-
6	32084485	C	T	missense_variant	*ATF6B*	33	het	1817/2607	1784/2103	595/700	R/Q	cGa/cAa	16/18	1388	NM_001136153.1	1.65E-04	2.08E-04	rs148987710
6	32130656	G	A	missense_variant	*PPT2*	22.7	het	1242/2044	838/909	280/302	G/S	Ggt/Agt	42987	9374	NM_001204103.1	NA	1.04E-04	-
6	32134378	C	T	missense_variant	*EGFL8*	27.1	het	286/1292	205/882	69/293	R/C	Cgc/Tgc	42803	80864	NM_030652.3	1.24E-04	2.08E-04	rs141849753
6	32149321	T	TC	frameshift_variant	*AGER*	NA	het	1100-1101/1393	1000-1001/1068	334/355	D/GX	gat/gGat	41526	177	NM_001206936.1	NA	2.08E-04	-
6	32151471	C	T	stop_gained	*AGER*	31	het	316/1490	216/1215	72/404	W/*	tgG/tgA	41343	177	NM_001136.4	NA	1.04E-04	rs200718197
6	32154455	G	A	stop_gained	*PBX2*	38	het	1397/3213	1126/1293	376/430	R/*	Cga/Tga	41494	5089	NM_002586.4	NA	1.04E-04	-
6	32155917	G	A	missense_variant	*PBX2*	24.8	het	831/3213	560/1293	187/430	T/M	aCg/aTg	42834	5089	NM_002586.4	6.59E-05	1.04E-04	rs149461273
6	32159646	GTC	G	frameshift_variant	*GPSM3*	NA	het	585-586/1485	183-184/483	61-62/160	QT/HX	caGAct/cact	41336	63940	NM_001276501.1	NA	1.04E-04	-
6	32164178	C	T	missense_variant	*NOTCH4*	36	het	5360/6745	5221/6012	1741/2003	A/T	Gct/Act	29/30	4855	NM_004557.3	6.81E-05	3.12E-04	-
6	32166872	C	T	missense_variant	*NOTCH4*	22.8	het	4505/6745	4366/6012	1456/2003	G/R	Ggg/Agg	24/30	4855	NM_004557.3	8.54E-06	1.04E-04	-
6	32168695	G	A	missense_variant	*NOTCH4*	21.3	het	4367/6745	4228/6012	1410/2003	R/C	Cgc/Tgc	23/30	4855	NM_004557.3	8.45E-06	1.04E-04	rs143103753
6	43752295	G	A	missense_variant	*VEGFA*	23.1	het	1733/3662	1235/1239	412/412	R/Q	cGg/cAg	42955	7422	NM_001025366.2	8.24E-06	1.04E-04	rs374253522
7	99943487	G	C	splice_acceptor_variant&non_coding_transcript_variant	*STAG3L5P-PVRIG2P-PILRB*	0.208	het	-/3683	-	-	-	-	-	101752399	NR_036569.1	NA	3.12E-04	rs2405449
7	99943487	G	C	splice_acceptor_variant&non_coding_transcript_variant	*STAG3L5P-PVRIG2P-PILRB*	0.208	het	-/3683	-	-	-	-	-	101752399	NR_036569.1	NA	3.12E-04	rs2405449
7	99956624	C	T	stop_gained	*PILRB*	14.32	het	676/1438	376/684	126/227	R/*	Cga/Tga	41308	29990	NM_178238.3	3.30E-05	1.04E-04	-
7	99995536	G	C	splice_donor_variant	*PILRA*	13.51	het	-/1316	-/912	-/303	-	-	-	29992	NM_013439.2	8.98E-04	8.32E-04	rs201649203
7	99995536	G	C	splice_donor_variant	*PILRA*	13.51	het	-/1316	-/912	-/303	-	-	-	29992	NM_013439.2	8.98E-04	8.32E-04	rs201649203
7	99995536	G	C	splice_donor_variant	*PILRA*	13.51	het	-/1316	-/912	-/303	-	-	-	29992	NM_013439.2	8.98E-04	8.32E-04	rs201649203
7	99997500	C	T	stop_gained	*PILRA*	15.05	het	803/1022	625/681	209/226	R/*	Cga/Tga	41430	29992	XM_005250301.1	9.06E-05	2.08E-04	rs374643830
7	99997500	C	T	stop_gained	*PILRA*	15.05	het	803/1022	625/681	209/226	R/*	Cga/Tga	41430	29992	XM_005250301.1	9.06E-05	2.08E-04	rs374643830
7	100004381	C	T	missense_variant	*ZCWPW1*	26.5	het	1486/2255	1109/1575	370/524	R/H	cGt/cAt	43085	55063	NM_001258008.1	4.14E-04	1.04E-04	rs201574533
7	100007051	C	T	missense_variant&splice_region_variant	*ZCWPW1*	23.5	het	1248/2255	871/1575	291/524	D/N	Gat/Aat	42994	55063	NM_001258008.1	6.04E-04	4.16E-04	rs199642636
7	100007051	C	T	missense_variant&splice_region_variant	*ZCWPW1*	23.5	het	1248/2255	871/1575	291/524	D/N	Gat/Aat	42994	55063	NM_001258008.1	6.04E-04	4.16E-04	rs199642636
7	100007051	C	T	missense_variant&splice_region_variant	*ZCWPW1*	23.5	het	1248/2255	871/1575	291/524	D/N	Gat/Aat	42994	55063	NM_001258008.1	6.04E-04	4.16E-04	rs199642636
7	100022652	C	T	splice_donor_variant	*ZCWPW1*	13.85	het	-/2255	-/1575	-/524	-	-	-	55063	NM_001258008.1	NA	1.04E-04	-
7	100071904	C	T	missense_variant	*TSC22D4*	21.2	het	1677/2383	922/1188	308/395	D/N	Gat/Aat	42799	81628	NM_030935.3	6.59E-04	7.28E-04	rs151058099
7	100071904	C	T	missense_variant	*TSC22D4*	21.2	het	1677/2383	922/1188	308/395	D/N	Gat/Aat	42799	81628	NM_030935.3	6.59E-04	7.28E-04	rs151058099
7	100071904	C	T	missense_variant	*TSC22D4*	21.2	het	1677/2383	922/1188	308/395	D/N	Gat/Aat	42799	81628	NM_030935.3	6.59E-04	7.28E-04	rs151058099
7	100071904	C	T	missense_variant	*TSC22D4*	21.2	het	1677/2383	922/1188	308/395	D/N	Gat/Aat	42799	81628	NM_030935.3	6.59E-04	7.28E-04	rs151058099
7	100084515	G	A	missense_variant	*NYAP1*	23.5	het	299/3584	140/2526	47/841	R/Q	cGg/cAg	42801	222950	NM_173564.3	2.40E-04	3.12E-04	rs202135611
7	100084527	C	T	missense_variant	*NYAP1*	36	het	311/3584	152/2526	51/841	S/L	tCg/tTg	42801	222950	NM_173564.3	2.54E-05	1.04E-04	-
7	100088158	G	A	missense_variant	*NYAP1*	21.7	het	2123/3584	1964/2526	655/841	R/Q	cGg/cAg	42862	222950	NM_173564.3	1.16E-04	3.12E-04	rs199612371
7	100088158	G	A	missense_variant	*NYAP1*	21.7	het	2123/3584	1964/2526	655/841	R/Q	cGg/cAg	42862	222950	NM_173564.3	1.16E-04	3.12E-04	rs199612371
7	104747653	G	A	missense_variant	*KMT2E*	23.2	het	3274/6715	2888/5577	963/1858	R/H	cGc/cAc	20/26	55904	NM_018682.3	4.13E-05	2.08E-04	rs370198555
7	104786961	G	C	missense_variant	*SRPK2*	25.8	het	914/3712	733/2067	245/688	P/A	Cct/Gct	42995	6733	NM_001278273.1	NA	1.04E-04	-
7	104801040	C	T	missense_variant	*SRPK2*	36	het	683/3712	502/2067	168/688	V/I	Gta/Ata	42964	6733	NM_001278273.1	8.24E-06	2.08E-04	-
7	104801040	C	T	missense_variant	*SRPK2*	36	het	683/3712	502/2067	168/688	V/I	Gta/Ata	42964	6733	NM_001278273.1	8.24E-06	2.08E-04	-
8	23056950	C	T	stop_gained	*TNFRSF10A*	12.69	het	949/1750	843/1407	281/468	W/*	tgG/tgA	41495	8797	NM_003844.3	NA	1.04E-04	-
8	23060160	C	G	splice_donor_variant	*TNFRSF10A*	15.19	het	-/1750	-/1407	-/468	-	-	-	8797	NM_003844.3	8.98E-04	5.20E-04	rs145440694
8	23060160	C	G	splice_donor_variant	*TNFRSF10A*	15.19	het	-/1750	-/1407	-/468	-	-	-	8797	NM_003844.3	8.98E-04	5.20E-04	rs145440694
8	23147516	G	A	missense_variant	*R3HCC1*	20.3	het	506/1625	506/1443	169/480	R/H	cGt/cAt	42864	203069	XM_005273427.1	1.64E-04	6.24E-04	-
8	23147516	G	A	missense_variant	*R3HCC1*	20.3	het	506/1625	506/1443	169/480	R/H	cGt/cAt	42864	203069	XM_005273427.1	1.64E-04	6.24E-04	-
8	23148976	G	A	missense_variant	*R3HCC1*	20.3	het	1032/1582	394/762	132/253	V/M	Gtg/Atg	42863	203069	NM_001136108.1	7.61E-05	1.04E-04	-
8	23156435	A	G	missense_variant	*LOXL2*	23.7	het	2498/3810	2159/2325	720/774	V/A	gTt/gCt	13/14	4017	NM_002318.2	5.19E-04	5.20E-04	rs148111808
8	23156435	A	G	missense_variant	*LOXL2*	23.7	het	2498/3810	2159/2325	720/774	V/A	gTt/gCt	13/14	4017	NM_002318.2	5.19E-04	5.20E-04	rs148111808
8	23159539	C	G	splice_donor_variant	*LOXL2*	13.49	het	-/3810	-/2325	-/774	-	-	-	4017	NM_002318.2	1.65E-05	1.04E-04	rs200988156
8	23167180	C	T	splice_donor_variant	*LOXL2*	26.5	het	-/3810	-/2325	-/774	-	-	-	4017	NM_002318.2	1.10E-04	2.08E-04	rs144719588
8	23174498	C	T	missense_variant	*LOXL2*	28.5	het	1939/3810	1600/2325	534/774	G/R	Gga/Aga	42992	4017	NM_002318.2	0.013	7.28E-04	rs149587418
8	23174498	C	T	missense_variant	*LOXL2*	28.5	het	1939/3810	1600/2325	534/774	G/R	Gga/Aga	42992	4017	NM_002318.2	0.013	7.28E-04	rs149587418
8	23174580	C	T	missense_variant	*LOXL2*	35	het	1857/3810	1518/2325	506/774	M/I	atG/atA	42992	4017	NM_002318.2	NA	1.04E-04	-
9	101817567	CA	C	frameshift_variant	*COL15A1*	NA	het	3455/5422	3106/4167	1036/1388	K/X	Aaa/aa	34/42	1306	NM_001855.4	8.24E-05	3.12E-04	-
9	101817567	CA	C	frameshift_variant	*COL15A1*	NA	het	3455/5422	3106/4167	1036/1388	K/X	Aaa/aa	34/42	1306	NM_001855.4	8.24E-05	3.12E-04	-
9	101824587	G	A	splice_donor_variant	*COL15A1*	21.1	het	-/5422	-/4167	-/1388	-	-	-	1306	NM_001855.4	8.24E-06	1.04E-04	-
9	101830877	G	A	missense_variant	*COL15A1*	20.10	het	4227/5422	3878/4167	1293/1388	G/D	gGc/gAc	41/42	1306	NM_001855.4	3.30E-04	7.28E-04	rs141581682
9	101830877	G	A	missense_variant	*COL15A1*	20.10	het	4227/5422	3878/4167	1293/1388	G/D	gGc/gAc	41/42	1306	NM_001855.4	3.30E-04	7.28E-04	rs141581682
9	107547873	G	A	missense_variant	*ABCA1*	25.7	het	6853/10503	6449/6786	2150/2261	P/L	cCg/cTg	49/50	19	NM_005502.3	4.12E-05	2.08E-04	rs369098049
9	107549164	T	A	missense_variant	*ABCA1*	31	het	6702/10503	6298/6786	2100/2261	T/S	Aca/Tca	47/50	19	NM_005502.3	NA	1.04E-04	-
9	107560803	C	T	missense_variant	*ABCA1*	29.5	het	5424/10503	5020/6786	1674/2261	V/I	Gta/Ata	37/50	19	NM_005502.3	6.92E-04	2.08E-04	rs138422574
9	107582258	T	C	missense_variant	*ABCA1*	31	het	3457/10503	3053/6786	1018/2261	D/G	gAt/gGt	21/50	19	NM_005502.3	4.37E-04	8.32E-04	rs140365800
9	107582258	T	C	missense_variant	*ABCA1*	31	het	3457/10503	3053/6786	1018/2261	D/G	gAt/gGt	21/50	19	NM_005502.3	4.37E-04	8.32E-04	rs140365800
9	107582258	T	C	missense_variant	*ABCA1*	31	het	3457/10503	3053/6786	1018/2261	D/G	gAt/gGt	21/50	19	NM_005502.3	4.37E-04	8.32E-04	rs140365800
9	107583723	G	A	missense_variant	*ABCA1*	24.7	het	3297/10503	2893/6786	965/2261	R/C	Cgc/Tgc	20/50	19	NM_005502.3	4.12E-05	1.04E-04	rs375095390
9	107586800	C	T	missense_variant	*ABCA1*	20.7	het	3006/10503	2602/6786	868/2261	E/K	Gag/Aag	18/50	19	NM_005502.3	1.43E-03	3.12E-04	rs35207495
9	107586800	C	T	missense_variant	*ABCA1*	20.7	het	3006/10503	2602/6786	868/2261	E/K	Gag/Aag	18/50	19	NM_005502.3	1.43E-03	3.12E-04	rs35207495
9	107599731	G	T	missense_variant	*ABCA1*	20.3	het	1576/10503	1172/6786	391/2261	A/D	gCc/gAc	18537	19	NM_005502.3	NA	1.04E-04	-
9	107646756	G	A	missense_variant	*ABCA1*	32	het	658/10503	254/6786	85/2261	P/L	cCg/cTg	18354	19	NM_005502.3	1.26E-03	5.20E-04	rs145183203
9	107646756	G	A	missense_variant	*ABCA1*	32	het	658/10503	254/6786	85/2261	P/L	cCg/cTg	18354	19	NM_005502.3	1.26E-03	5.20E-04	rs145183203
10	24810825	G	A	missense_variant	*KIAA1217*	32	het	2992/6123	2183/3795	728/1264	R/H	cGc/cAc	13/19	56243	NM_001098500.1	2.31E-04	4.16E-04	rs141531788
10	24810825	G	A	missense_variant	*KIAA1217*	32	het	2992/6123	2183/3795	728/1264	R/H	cGc/cAc	13/19	56243	NM_001098500.1	2.31E-04	4.16E-04	rs141531788
10	24816899	A	G	missense_variant	*KIAA1217*	21.5	het	3502/6123	2693/3795	898/1264	N/S	aAt/aGt	15/19	56243	NM_001098500.1	4.94E-05	2.08E-04	rs375000847
10	24816899	A	G	missense_variant	*KIAA1217*	21.5	het	3502/6123	2693/3795	898/1264	N/S	aAt/aGt	15/19	56243	NM_001098500.1	4.94E-05	2.08E-04	rs375000847
10	24816911	A	G	missense_variant	*KIAA1217*	20.5	het	3514/6123	2705/3795	902/1264	Y/C	tAt/tGt	15/19	56243	NM_001098500.1	4.86E-04	5.20E-04	rs138435732
10	24816944	A	C	missense_variant	*KIAA1217*	28.3	het	3547/6123	2738/3795	913/1264	E/A	gAa/gCa	15/19	56243	NM_001098500.1	5.77E-05	1.04E-04	rs374904740
10	24816976	A	C	missense_variant	*KIAA1217*	22.1	het	3579/6123	2770/3795	924/1264	N/H	Aat/Cat	15/19	56243	NM_001098500.1	NA	1.04E-04	-
10	24874897	A	T	missense_variant	*ARHGAP21*	23.7	het	4808/7167	4321/5877	1441/1958	F/I	Ttt/Att	26/26	57584	NM_020824.3	3.30E-05	2.08E-04	-
10	24874897	A	T	stop_gained	*ARHGAP21*	23.7	het	4660/4919	4215/4374	1405/1457	Y/*	taT/taA	25/25	57584	XM_005252546.1	3.30E-05	2.08E-04	-

10	24885785	C	T	missense_variant	*ARHGAP21*	30	het	3848/7167	3361/5877	1121/1958	G/S	Ggc/Agc	17/26	57584	NM_020824.3	6.45E-03	3.12E-04	rs111419738
10	24885785	C	T	missense_variant	*ARHGAP21*	30	het	3848/7167	3361/5877	1121/1958	G/S	Ggc/Agc	17/26	57584	NM_020824.3	6.45E-03	3.12E-04	rs111419738
10	24885785	C	T	missense_variant	*ARHGAP21*	30	het	3848/7167	3361/5877	1121/1958	G/S	Ggc/Agc	17/26	57584	NM_020824.3	6.45E-03	3.12E-04	rs111419738
10	24889799	G	C	missense_variant	*ARHGAP21*	22.1	het	3395/7167	2908/5877	970/1958	L/V	Ctt/Gtt	14/26	57584	NM_020824.3	4.86E-04	3.12E-04	rs144895449
10	24910072	G	A	missense_variant	*ARHGAP21*	24.7	het	1239/7167	752/5877	251/1958	P/L	cCt/cTt	43004	57584	NM_020824.3	1.65E-05	1.04E-04	-
10	124189247	CT	C	frameshift_variant	*PLEKHA1*	NA	het	1258/3881	1009/1215	337/404	F/X	Ttt/tt	41619	59338	NM_001001974.2	1.89E-04	6.24E-04	-
10	124189247	CT	C	frameshift_variant	*PLEKHA1*	NA	het	1258/3881	1009/1215	337/404	F/X	Ttt/tt	41619	59338	NM_001001974.2	1.89E-04	6.24E-04	-
10	124189247	CT	C	frameshift_variant	*PLEKHA1*	NA	het	1258/3881	1009/1215	337/404	F/X	Ttt/tt	41619	59338	NM_001001974.2	1.89E-04	6.24E-04	-
10	124189247	CT	C	frameshift_variant	*PLEKHA1*	NA	het	1258/3881	1009/1215	337/404	F/X	Ttt/tt	41619	59338	NM_001001974.2	1.89E-04	6.24E-04	-
10	124249143	G	A	splice_donor_variant	*HTRA1*	21.9	het	-/2120	-/1443	-/480	-	-	-	5654	NM_002775.4	8.24E-06	1.04E-04	rs370332995
10	124266333	C	T	stop_gained	*HTRA1*	40	het	1032/2120	904/1443	302/480	R/*	Cga/Tga	41372	5654	NM_002775.4	1.65E-05	1.04E-04	rs113993970
12	56075710	C	T	missense_variant	*METTL7B*	23.4	het	381/1506	172/735	58/244	L/F	Ctc/Ttc	42737	196410	NM_152637.2	NA	2.08E-04	-
12	56077768	C	T	stop_gained	*METTL7B*	18.73	het	879/1506	670/735	224/244	R/*	Cga/Tga	41306	196410	NM_152637.2	4.00E-03	3.12E-04	rs115687886
12	56077768	C	T	stop_gained	*METTL7B*	18.73	het	879/1506	670/735	224/244	R/*	Cga/Tga	41306	196410	NM_152637.2	4.00E-03	3.12E-04	rs115687886
12	56077768	C	T	stop_gained	*METTL7B*	18.73	het	879/1506	670/735	224/244	R/*	Cga/Tga	41306	196410	NM_152637.2	4.00E-03	3.12E-04	rs115687886
12	56079053	G	A	missense_variant	*ITGA7*	43	het	3435/4136	3215/3426	1072/1141	A/V	gCg/gTg	25/25	3679	NM_001144996.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	stop_gained	*ITGA7*	43	het	3801/4502	3448/3489	1150/1162	R/*	Cga/Tga	27/27	3679	XM_005268841.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	missense_variant	*ITGA7*	43	het	3435/4136	3215/3426	1072/1141	A/V	gCg/gTg	25/25	3679	NM_001144996.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	stop_gained	*ITGA7*	43	het	3801/4502	3448/3489	1150/1162	R/*	Cga/Tga	27/27	3679	XM_005268841.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	missense_variant	*ITGA7*	43	het	3435/4136	3215/3426	1072/1141	A/V	gCg/gTg	25/25	3679	NM_001144996.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	stop_gained	*ITGA7*	43	het	3801/4502	3448/3489	1150/1162	R/*	Cga/Tga	27/27	3679	XM_005268841.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	missense_variant	*ITGA7*	43	het	3435/4136	3215/3426	1072/1141	A/V	gCg/gTg	25/25	3679	NM_001144996.1	9.23E-04	8.32E-04	rs139136931
12	56079053	G	A	stop_gained	*ITGA7*	43	het	3801/4502	3448/3489	1150/1162	R/*	Cga/Tga	27/27	3679	XM_005268841.1	9.23E-04	8.32E-04	rs139136931
12	56090189	T	G	missense_variant	*ITGA7*	32	het	1997/4136	1777/3426	593/1141	I/L	Att/Ctt	13/25	3679	NM_001144996.1	2.89E-04	6.24E-04	rs200065922
12	56090189	T	G	missense_variant	*ITGA7*	32	het	1997/4136	1777/3426	593/1141	I/L	Att/Ctt	13/25	3679	NM_001144996.1	2.89E-04	6.24E-04	rs200065922
12	56094076	G	A	missense_variant	*ITGA7*	20.7	het	992/4136	772/3426	258/1141	L/F	Ctc/Ttc	42880	3679	NM_001144996.1	4.15E-05	2.08E-04	-
12	56095165	C	A	stop_gained&splice_region_variant	*ITGA7*	13.11	het	157/3661	121/3135	41/1044	E/*	Gaa/Taa	41357	3679	NM_001144997.1	1.36E-05	2.08E-04	-
12	56112882	C	T	stop_gained	*BLOC1S1*	26.6	het	244/578	226/462	76/153	Q/*	Cag/Tag	41336	2647	NM_001487.3	NA	1.04E-04	-
12	56142589	G	A	missense_variant	*GDF11*	24.5	het	702/4260	665/1224	222/407	R/H	cGc/cAc	42769	10220	NM_005811.3	NA	2.08E-04	-
12	56212958	G	C	splice_donor_variant	*ORMDL2*	16.82	het	-/1334	-/462	-/153	-	-	-	29095	NM_014182.4	3.30E-05	2.08E-04	-
12	56212958	G	C	splice_donor_variant	*ORMDL2*	16.82	het	-/1334	-/462	-/153	-	-	-	29095	NM_014182.4	3.30E-05	2.08E-04	-
12	56212958	G	GT	splice_donor_variant	*ORMDL2*	NA	het	-/1334	-/462	-/153	-	-	-	29095	NM_014182.4	NA	1.04E-04	-
12	56213223	A	G	missense_variant	*ORMDL2*	21.5	het	364/1334	272/462	91/153	Y/C	tAt/tGt	42798	29095	NM_014182.4	1.65E-05	1.04E-04	-
12	112147438	C	T	missense_variant	*ACAD10*	22	het	840/4107	640/3273	214/1090	L/F	Ctt/Ttt	42877	80724	NM_001136538.1	4.20E-04	2.08E-04	rs199662727
12	112153654	G	A	missense_variant	*ACAD10*	21.6	het	1173/4107	973/3273	325/1090	G/R	Ggg/Agg	42969	80724	NM_001136538.1	7.41E-05	2.08E-04	-
12	112186211	T	C	missense_variant	*ACAD10*	23.8	het	2869/4107	2669/3273	890/1090	V/A	gTt/gCt	18/22	80724	NM_001136538.1	3.46E-04	4.16E-04	rs374524601
12	112186211	T	C	missense_variant	*ACAD10*	23.8	het	2869/4107	2669/3273	890/1090	V/A	gTt/gCt	18/22	80724	NM_001136538.1	3.46E-04	4.16E-04	rs374524601
12	112194215	G	A	missense_variant	*ACAD10*	24.8	het	3411/4107	3211/3273	1071/1090	D/N	Gac/Aac	22/22	80724	NM_001136538.1	2.88E-04	5.20E-04	rs142030532
12	112235917	C	A	stop_gained	*ALDH2*	13.03	het	1215/2060	1119/1554	373/517	Y/*	taC/taA	41559	217	NM_000690.3	NA	1.04E-04	-
12	112241766	G	A	missense_variant	*ALDH2*	35	het	1606/2060	1510/1554	504/517	E/K	Gaa/Aaa	43082	217	NM_000690.3	0.017	1.04E-04	rs671
13	31722129	G	T	missense_variant	*HSPH1*	22.4	het	1743/3668	1345/2577	449/858	P/T	Ccc/Acc	43026	10808	NM_006644.2	1.67E-03	4.16E-04	rs148641546
13	31722186	T	G	missense_variant	*HSPH1*	28.6	het	1686/3668	1288/2577	430/858	K/Q	Aaa/Caa	43026	10808	NM_006644.2	1.65E-05	1.04E-04	-
13	31898021	C	G	missense_variant	*B3GALTL*	28.6	het	1428/4215	1318/1497	440/498	L/V	Ctc/Gtc	14/15	145173	NM_194318.3	9.06E-05	2.08E-04	rs142935066
13	31898021	C	G	missense_variant	*B3GALTL*	28.6	het	1428/4215	1318/1497	440/498	L/V	Ctc/Gtc	14/15	145173	NM_194318.3	9.06E-05	2.08E-04	rs142935066
13	31903713	G	T	missense_variant	*B3GALTL*	25.6	het	1515/4215	1405/1497	469/498	D/Y	Gat/Tat	15/15	145173	NM_194318.3	1.07E-04	3.12E-04	rs141036237
14	69028735	C	T	stop_gained	*RAD51B*	5.376	het	1134/3051	1057/1119	353/372	Q/*	Cag/Tag	41588	5890	XM_005267961.1	NA	4.16E-04	rs74821294
16	56901103	G	A	missense_variant	*SLC12A3*	25.1	het	433/5567	404/3093	135/1030	R/H	cGc/cAc	42792	6559	NM_000339.2	NA	2.08E-04	-
16	56913524	C	T	missense_variant	*SLC12A3*	34	het	1435/5567	1406/3093	469/1030	A/V	gCc/gTc	43065	6559	NM_000339.2	7.41E-05	1.04E-04	rs139743444
16	56921879	G	A	missense_variant	*SLC12A3*	21.6	het	2250/5567	2221/3093	741/1030	G/R	Ggg/Agg	18/26	6559	NM_000339.2	4.04E-04	7.28E-04	rs138977195
16	56921879	G	A	missense_variant	*SLC12A3*	21.6	het	2250/5567	2221/3093	741/1030	G/R	Ggg/Agg	18/26	6559	NM_000339.2	4.04E-04	7.28E-04	rs138977195
16	56936421	G	T	splice_donor_variant	*SLC12A3*	16.11	het	-/5567	-/3093	-/1030	-	-	-	6559	NM_000339.2	1.73E-04	5.20E-04	rs199974259
16	56936421	G	T	splice_donor_variant	*SLC12A3*	16.11	het	-/5567	-/3093	-/1030	-	-	-	6559	NM_000339.2	1.73E-04	5.20E-04	rs199974259
16	56936421	G	T	splice_donor_variant	*SLC12A3*	16.11	het	-/5567	-/3093	-/1030	-	-	-	6559	NM_000339.2	1.73E-04	5.20E-04	rs199974259
16	56936421	G	T	splice_donor_variant	*SLC12A3*	16.11	het	-/5567	-/3093	-/1030	-	-	-	6559	NM_000339.2	1.73E-04	5.20E-04	rs199974259
16	56969214	T	C	missense_variant	*HERPUD1*	32	het	370/1919	215/1173	72/390	L/S	tTg/tCg	42774	9709	NM_001010989.2	4.12E-05	1.04E-04	-
16	56970635	C	T	missense_variant	*HERPUD1*	24.2	het	489/1919	334/1173	112/390	R/W	Cgg/Tgg	42833	9709	NM_001010989.2	2.47E-05	1.04E-04	rs368534600
16	57003374	C	T	stop_gained	*CETP*	18.3	het	367/1717	310/1482	104/493	Q/*	Cag/Tag	41348	1071	NM_000078.2	8.24E-06	1.04E-04	-
16	57017318	C	T	stop_gained	*CETP*	15.73	het	1459/1717	1402/1482	468/493	R/*	Cga/Tga	15/16	1071	NM_000078.2	2.47E-05	1.04E-04	-
16	75148093	C	T	stop_gained	*LDHD*	12.31	het	721/2072	669/1524	223/507	W/*	tgG/tgA	41435	197257	NM_153486.3	1.81E-04	6.24E-04	rs139739338
16	75148093	C	T	stop_gained	*LDHD*	12.31	het	721/2072	669/1524	223/507	W/*	tgG/tgA	41435	197257	NM_153486.3	1.81E-04	6.24E-04	rs139739338
16	75258615	G	A	missense_variant	*CTRB1*	20.7	het	665/872	643/792	215/263	G/S	Ggc/Agc	42923	1504	NM_001906.4	9.88E-05	1.04E-04	rs375112297
16	75269046	C	T	missense_variant	*BCAR1*	26.2	het	2173/3516	1889/2751	630/916	R/Q	cGg/cAg	42894	9564	NM_001170714.1	8.85E-04	9.36E-04	rs139270175
16	75269046	C	T	missense_variant	*BCAR1*	26.2	het	2173/3516	1889/2751	630/916	R/Q	cGg/cAg	42894	9564	NM_001170714.1	8.85E-04	9.36E-04	rs139270175
16	75298398	T	G	start_lost	*BCAR1*	9.407	het	285/3516	309361	1/916	M/L	Atg/Ctg	41312	9564	NM_001170714.1	1.53E-03	3.12E-04	rs143615054
16	75298398	T	G	start_lost	*BCAR1*	9.407	het	285/3516	309361	1/916	M/L	Atg/Ctg	41312	9564	NM_001170714.1	1.53E-03	3.12E-04	rs143615054
17	26669403	G	C	missense_variant	*TNFAIP1*	34	het	1098/3809	649/951	217/316	G/R	Ggc/Cgc	42893	7126	NM_021137.4	1.81E-04	1.04E-04	rs138622447
17	26671495	C	T	missense_variant	*TNFAIP1*	20.10	het	1269/3809	820/951	274/316	R/C	Cgc/Tgc	42923	7126	NM_021137.4	2.47E-05	2.08E-04	rs200262864
17	26671495	C	T	missense_variant	*TNFAIP1*	20.10	het	1269/3809	820/951	274/316	R/C	Cgc/Tgc	42923	7126	NM_021137.4	2.47E-05	2.08E-04	rs200262864
17	26671495	C	T	missense_variant	*TNFAIP1*	20.10	het	1269/3809	820/951	274/316	R/C	Cgc/Tgc	42923	7126	NM_021137.4	2.47E-05	2.08E-04	rs200262864
17	26694877	G	A	missense_variant	*VTN*	20.8	het	1332/1678	1183/1437	395/478	R/W	Cgg/Tgg	42924	7448	NM_000638.3	2.47E-05	1.04E-04	rs368991682
17	26695950	CTGCAT	C	frameshift_variant	*VTN*	NA	het	913-917/1678	764-768/1437	255-256/478	DA/X	gATGCA/g	41401	7448	NM_000638.3	1.65E-05	1.04E-04	-
17	26695998	G	A	stop_gained	*VTN*	43	het	870/1678	721/1437	241/478	R/*	Cga/Tga	41401	7448	NM_000638.3	4.12E-05	2.08E-04	rs199987147
17	26696531	G	A	stop_gained	*VTN*	42	het	675/1678	526/1437	176/478	R/*	Cga/Tga	41340	7448	NM_000638.3	2.47E-05	1.04E-04	-
17	26696772	G	GC	frameshift_variant	*VTN*	NA	het	433-434/1678	284-285/1437	95/478	G/GX	ggc/ggGc	41340	7448	NM_000638.3	5.60E-04	4.16E-04	-
17	26696772	G	GC	frameshift_variant	*VTN*	NA	het	433-434/1678	284-285/1437	95/478	G/GX	ggc/ggGc	41340	7448	NM_000638.3	5.60E-04	4.16E-04	-
17	26696772	G	GC	frameshift_variant	*VTN*	NA	het	433-434/1678	284-285/1437	95/478	G/GX	ggc/ggGc	41340	7448	NM_000638.3	5.60E-04	4.16E-04	-
17	26708221	G	A	splice_acceptor_variant	*SARM1*	17.95	het	-/7301	-/2173	-/724	-	-	-	23098	NM_015077.3	2.47E-05	2.08E-04	rs369515982
17	26824148	G	T	missense_variant	*SLC13A2*	21.8	het	1854/2505	1787/1926	596/641	G/V	gGa/gTa	43081	9058	NM_001145975.1	NA	1.04E-04	-
17	79427598	G	A	missense_variant	*BAHCC1*	24.3	het	5897/10171	5897/7656	1966/2551	R/Q	cGg/cAg	24/27	57597	NM_001080519.2	1.08E-04	1.04E-04	rs200336497
17	79428068	G	A	missense_variant	*BAHCC1*	22.5	het	6208/10171	6208/7656	2070/2551	G/S	Ggc/Agc	25/27	57597	NM_001080519.2	NA	2.08E-04	rs199525294
17	79428068	G	A	missense_variant	*BAHCC1*	22.5	het	6208/10171	6208/7656	2070/2551	G/S	Ggc/Agc	25/27	57597	NM_001080519.2	NA	2.08E-04	rs199525294
17	79429896	G	A	missense_variant	*BAHCC1*	27.5	het	7343/10171	7343/7656	2448/2551	R/H	cGc/cAc	26/27	57597	NM_001080519.2	2.48E-04	6.24E-04	rs199906132
17	79430779	C	T	missense_variant	*BAHCC1*	36	het	7592/10171	7592/7656	2531/2551	A/V	gCg/gTg	27/27	57597	NM_001080519.2	NA	1.04E-04	-
17	79430806	G	A	missense_variant	*BAHCC1*	32	het	7619/10171	7619/7656	2540/2551	R/H	cGc/cAc	27/27	57597	NM_001080519.2	1.01E-04	3.12E-04	rs375282374

17	79495687	C	T	stop_gained	*FSCN2*	38	het	271/1742	130/1551	44/516	Q/*	Cag/Tag	41278	25794	NM_001077182.2	1.66E-04	3.12E-04	rs200925863
17	79495687	C	T	stop_gained	*FSCN2*	38	het	271/1742	130/1551	44/516	Q/*	Cag/Tag	41278	25794	NM_001077182.2	1.66E-04	3.12E-04	rs200925863
17	79496003	G	A	missense_variant	*FSCN2*	29	het	587/1742	446/1551	149/516	R/Q	cGg/cAg	42740	25794	NM_001077182.2	5.38E-04	7.28E-04	rs181961770
17	79496003	G	A	missense_variant	*FSCN2*	29	het	587/1742	446/1551	149/516	R/Q	cGg/cAg	42740	25794	NM_001077182.2	5.38E-04	7.28E-04	rs181961770
17	79580404	T	C	missense_variant	*NPLOC4*	23.8	het	508/4381	326/1827	109/608	E/G	gAg/gGg	42842	55666	NM_017921.2	8.27E-05	3.12E-04	rs367985830
17	79650146	T	C	missense_variant	*ARL16*	25.4	het	302/1207	203/594	68/197	N/S	aAt/aGt	42799	339231	NM_001040025.1	8.27E-06	3.12E-04	rs370905325
17	79650146	T	C	missense_variant	*ARL16*	25.4	het	302/1207	203/594	68/197	N/S	aAt/aGt	42799	339231	NM_001040025.1	8.27E-06	3.12E-04	rs370905325
17	79660595	T	C	missense_variant	*HGS*	23.2	het	860/2972	725/2334	242/777	L/P	cTg/cCg	43000	9146	NM_004712.4	7.09E-04	5.20E-04	rs200174222
17	79660595	T	C	missense_variant	*HGS*	23.2	het	860/2972	725/2334	242/777	L/P	cTg/cCg	43000	9146	NM_004712.4	7.09E-04	5.20E-04	rs200174222
17	79660609	C	G	missense_variant&splice_region_variant	*HGS*	24.8	het	874/2972	739/2334	247/777	Q/E	Cag/Gag	43000	9146	NM_004712.4	8.33E-04	8.32E-04	rs145607073
17	79660609	C	G	missense_variant&splice_region_variant	*HGS*	24.8	het	874/2972	739/2334	247/777	Q/E	Cag/Gag	43000	9146	NM_004712.4	8.33E-04	8.32E-04	rs145607073
17	79660609	C	G	missense_variant&splice_region_variant	*HGS*	24.8	het	874/2972	739/2334	247/777	Q/E	Cag/Gag	43000	9146	NM_004712.4	8.33E-04	8.32E-04	rs145607073
17	79663495	A	T	missense_variant	*HGS*	26.8	het	1637/2972	1502/2334	501/777	E/V	gAg/gTg	16/22	9146	NM_004712.4	NA	2.08E-04	-
17	79674182	A	G	missense_variant	*MRPL12*	25.2	het	641/1015	509/597	170/198	Q/R	cAg/cGg	42860	6182	NM_002949.3	1.65E-05	2.08E-04	-
17	79674182	A	G	missense_variant	*MRPL12*	25.2	het	641/1015	509/597	170/198	Q/R	cAg/cGg	42860	6182	NM_002949.3	1.65E-05	2.08E-04	-
19	990236	G	A	missense_variant	*WDR18*	21.9	het	495/1551	469/1299	157/432	D/N	Gac/Aac	42835	57418	NM_024100.3	6.65E-05	4.16E-04	-
19	990294	C	T	missense_variant	*WDR18*	25.7	het	553/1551	527/1299	176/432	T/M	aCg/aTg	42835	57418	NM_024100.3	2.49E-05	1.04E-04	-
19	990861	G	GTC	frameshift_variant	*WDR18*	NA	het	633-634/1551	607-608/1299	203/432	V/VX	gtc/gTCtc	41403	57418	NM_024100.3	8.26E-06	1.04E-04	-
19	1036553	G	A	missense_variant	*CNN2*	20.5	het	1009/2458	646/930	216/309	A/T	Gcc/Acc	42893	1265	NM_004368.2	8.24E-06	1.04E-04	-
19	1051006	G	A	missense_variant	*ABCA7*	33	het	2870/6819	2639/6441	880/2146	R/Q	cGg/cAg	19/47	10347	NM_019112.3	1.27E-03	6.24E-04	rs143718918
19	1051584	AG	A	frameshift_variant&splice_region_variant	*ABCA7*	NA	het	3193/6819	2962/6441	988/2146	G/X	Ggt/gt	21/47	10347	NM_019112.3	8.30E-06	1.04E-04	rs34430899
19	1055907	CT	C	frameshift_variant&splice_region_variant	*ABCA7*	NA	het	4439/6819	4208/6441	1403/2146	L/X	cTg/cg	31/47	10347	NM_019112.3	1.05E-03	9.36E-04	-
19	1055907	CT	C	frameshift_variant&splice_region_variant	*ABCA7*	NA	het	4439/6819	4208/6441	1403/2146	L/X	cTg/cg	31/47	10347	NM_019112.3	1.05E-03	9.36E-04	-
19	1055907	CT	C	frameshift_variant&splice_region_variant	*ABCA7*	NA	het	4439/6819	4208/6441	1403/2146	L/X	cTg/cg	31/47	10347	NM_019112.3	1.05E-03	9.36E-04	-
19	1055907	CT	C	frameshift_variant&splice_region_variant	*ABCA7*	NA	het	4439/6819	4208/6441	1403/2146	L/X	cTg/cg	31/47	10347	NM_019112.3	1.05E-03	9.36E-04	-
19	1091865	G	A	stop_gained	*POLR2E*	36	het	351/2866	274/633	92/210	Q/*	Cag/Tag	41340	5434	NM_002695.3	NA	2.08E-04	rs371458354
19	1091865	G	A	stop_gained	*POLR2E*	36	het	351/2866	274/633	92/210	Q/*	Cag/Tag	41340	5434	NM_002695.3	NA	2.08E-04	rs371458354
19	1109292	C	A	missense_variant&splice_region_variant	*SBNO2*	20.39	het	3253/4593	3176/3930	1059/1309	R/L	cGg/cTg	26/29	22904	NM_001100122.1	1.27E-04	1.04E-04	rs201689400
19	1119113	A	T	missense_variant	*SBNO2*	21.1	het	1330/4593	1253/3930	418/1309	M/K	aTg/aAg	43068	22904	NM_001100122.1	NA	1.04E-04	-
19	5714234	C	T	missense_variant	*LONP1*	28.4	het	319/2919	286/2688	96/895	A/T	Gcc/Acc	42813	9361	NM_001276479.1	5.27E-04	7.28E-04	rs148374055
19	5714234	C	T	missense_variant	*LONP1*	28.4	het	319/2919	286/2688	96/895	A/T	Gcc/Acc	42813	9361	NM_001276479.1	5.27E-04	7.28E-04	rs148374055
19	5714234	C	T	missense_variant	*LONP1*	28.4	het	319/2919	286/2688	96/895	A/T	Gcc/Acc	42813	9361	NM_001276479.1	5.27E-04	7.28E-04	rs148374055
19	5754142	G	C	splice_acceptor_variant	*CATSPERD*	7.033	het	-/2513	-/2397	-/798	-	-	-	257062	NM_152784.3	1.66E-05	1.04E-04	-
19	5831420	C	T	splice_acceptor_variant	*FUT6*	2.963	het	-/2651	-/1074	-/357	-	-	-	2528	XM_005259527.1	3.30E-05	3.12E-04	-
19	5831420	C	T	splice_acceptor_variant	*FUT6*	2.963	het	-/2651	-/1074	-/357	-	-	-	2528	XM_005259527.1	3.30E-05	3.12E-04	-
19	5832098	C	T	missense_variant	*FUT6*	22.8	het	1676/3137	481/1080	161/359	D/N	Gac/Aac	42797	2528	NM_000150.2	1.65E-05	1.04E-04	-
19	5893192	TAC	T	frameshift_variant	*NDUFA11*	NA	het	726-727/2633	421-422/687	141/228	V/X	GTa/a	41367	126328	NM_001193375.1	NA	2.08E-04	-
19	5924893	G	A	missense_variant	*RANBP3*	20.3	het	1153/3387	926/1689	309/562	A/V	gCc/gTc	43056	8498	NM_003624.2	6.46E-03	3.12E-04	rs10417885
19	6693427	T	C	missense_variant	*C3*	20.7	het	3288/5101	3226/4992	1076/1663	T/A	Acc/Gcc	25/41	718	NM_000064.2	NA	1.04E-04	-
19	6732550	T	A	missense_variant	*GPR108*	21.2	het	990/2076	944/1632	315/543	Y/F	tAc/tTc	43057	56927	NM_001080452.1	1.61E-03	8.32E-04	rs191648389
19	6732550	T	A	missense_variant	*GPR108*	21.2	hom	990/2076	944/1632	315/543	Y/F	tAc/tTc	43057	56927	NM_001080452.1	1.61E-03	8.32E-04	rs191648389
19	45323961	G	A	splice_acceptor_variant	*BCAM*	11.34	het	-/2453	-/1887	-/628	-	-	-	4059	NM_005581.4	NA	2.08E-04	-
19	45323961	G	A	splice_acceptor_variant	*BCAM*	11.34	het	-/2453	-/1887	-/628	-	-	-	4059	NM_005581.4	NA	2.08E-04	-
19	45381856	AC	A	frameshift_variant	*PVRL2*	NA	het	1810/2151	1420/1440	474/479	R/X	Cgg/gg	41430	5819	NM_002856.2	NA	1.04E-04	-
19	45515404	C	T	missense_variant	*RELB*	25	het	505/2279	374/1740	125/579	P/L	cCg/cTg	42837	5971	NM_006509.3	8.66E-06	1.04E-04	-
19	45815010	A	T	missense_variant	*CKM*	25.4	het	825/1658	650/1146	217/381	I/N	aTc/aAc	42863	1158	NM_001824.4	1.66E-04	1.04E-04	rs373872310
19	45815121	G	A	missense_variant	*CKM*	25.6	het	714/1658	539/1146	180/381	T/M	aCg/aTg	42863	1158	NM_001824.4	1.48E-04	2.08E-04	rs145987658
19	45815121	G	A	missense_variant	*CKM*	25.6	het	714/1658	539/1146	180/381	T/M	aCg/aTg	42863	1158	NM_001824.4	1.48E-04	2.08E-04	rs145987658
19	45818809	C	T	missense_variant	*CKM*	20.8	het	570/1658	395/1146	132/381	R/H	cGc/cAc	42833	1158	NM_001824.4	2.14E-04	2.08E-04	rs147574145
19	45852852	A	C	missense_variant	*KLC3*	21.1	het	1250/1793	1135/1515	379/504	N/H	Aac/Cac	42960	147700	NM_177417.2	1.58E-03	3.12E-04	rs146271778
19	45852852	A	C	missense_variant	*KLC3*	21.1	het	1250/1793	1135/1515	379/504	N/H	Aac/Cac	42960	147700	NM_177417.2	1.58E-03	3.12E-04	rs146271778
19	45856370	C	T	missense_variant	*ERCC2*	25.2	het	1849/2568	1802/2283	601/760	R/Q	cGg/cAg	19/23	2068	NM_000400.3	1.73E-04	2.08E-04	rs140522180
19	45860760	C	T	missense_variant	*ERCC2*	21	het	1396/2568	1349/2283	450/760	R/H	cGt/cAt	14/23	2068	NM_000400.3	2.14E-04	3.12E-04	rs146632315
19	45867532	C	T	missense_variant	*ERCC2*	27.1	het	823/2568	776/2283	259/760	C/Y	tGc/tAc	43001	2068	NM_000400.3	5.77E-05	5.20E-04	rs370454709
19	45868311	G	A	stop_gained	*ERCC2*	38	het	513/2568	466/2283	156/760	R/*	Cga/Tga	41447	2068	NM_000400.3	1.65E-05	1.04E-04	rs151235136
19	45868349	C	T	missense_variant	*ERCC2*	28.3	het	475/2568	428/2283	143/760	R/Q	cGg/cAg	42909	2068	NM_000400.3	1.32E-04	1.04E-04	rs150000483
19	45899469	G	A	missense_variant	*PPP1R13L*	24.4	het	939/3122	860/2487	287/828	P/L	cCt/cTt	42899	10848	NM_001142502.1	2.88E-04	3.12E-04	rs142842464
19	45909724	C	CT	frameshift_variant	*PPP1R13L*	NA	het	63-64/3299	14-15/2697	5/898	K/KX	aag/aaAg	41286	10848	XM_005258424.1	NA	1.04E-04	-
19	45911461	C	T	missense_variant	*CD3EAP*	21	het	723/3286	235/1533	79/510	R/C	Cgc/Tgc	42797	10849	NM_012099.1	3.30E-05	1.04E-04	rs143485921
19	45912181	G	A	missense_variant	*CD3EAP*	21	het	1443/3286	955/1533	319/510	E/K	Gag/Aag	42797	10849	NM_012099.1	1.16E-03	6.24E-04	rs143443617
20	44515299	C	T	missense_variant	*SPATA25*	23.6	het	550/719	541/684	181/227	A/T	Gct/Act	42768	128497	NM_080608.3	2.39E-04	6.24E-04	rs200024163
20	44519152	AC	A	frameshift_variant	*NEURL2*	NA	het	774/1454	478/786	160/261	V/X	Gtg/tg	41275	140825	NM_001278535.1	NA	1.04E-04	-
20	44521386	C	T	missense_variant	*CTSA*	36	het	895/2241	521/1497	174/498	A/V	gCc/gTc	42901	5476	NM_000308.2	9.88E-05	2.08E-04	rs139760741
20	44531218	A	G	missense_variant	*PLTP*	27.8	het	980/1815	683/1197	228/398	L/S	tTg/tCg	42992	5360	NM_001242920.1	1.98E-04	4.16E-04	rs373772930
20	44531218	A	G	missense_variant	*PLTP*	27.8	het	980/1815	683/1197	228/398	L/S	tTg/tCg	42992	5360	NM_001242920.1	1.98E-04	4.16E-04	rs373772930
20	44534995	C	T	missense_variant	*PLTP*	24.3	het	629/1815	332/1197	111/398	R/H	cGc/cAc	42900	5360	NM_001242920.1	1.32E-04	1.04E-04	-
20	44569202	G	A	missense_variant	*PCIF1*	25.4	het	652/2697	338/2115	113/704	R/Q	cGg/cAg	42872	63935	NM_022104.3	6.51E-04	5.20E-04	rs188509769
20	44577623	T	C	missense_variant	*ZNF335*	22.8	het	4121/4452	3998/4029	1333/1342	E/G	gAg/gGg	28/28	63925	NM_022095.3	9.06E-05	3.12E-04	rs200635860
20	44578900	T	C	missense_variant	*ZNF335*	20.9	het	3568/4452	3445/4029	1149/1342	I/V	Atc/Gtc	22/28	63925	NM_022095.3	NA	4.16E-04	rs143113106
20	44587908	T	A	missense_variant	*ZNF335*	28	het	2308/4452	2185/4029	729/1342	I/F	Att/Ttt	15/28	63925	NM_022095.3	8.41E-04	7.28E-04	rs190178539
20	44589092	G	A	missense_variant	*ZNF335*	20.6	het	1978/4452	1855/4029	619/1342	R/C	Cgc/Tgc	13/28	63925	NM_022095.3	9.88E-05	3.12E-04	rs144619729
20	44592204	G	C	missense_variant	*ZNF335*	20.5	het	1564/4452	1441/4029	481/1342	R/G	Cgc/Ggc	43006	63925	NM_022095.3	9.06E-05	5.20E-04	rs141481390
20	44638599	T	A	missense_variant	*MMP9*	22.7	het	252/2336	233/2124	78/707	L/Q	cTg/cAg	42779	4318	NM_004994.2	NA	1.04E-04	-
20	44638668	TC	T	frameshift_variant	*MMP9*	NA	het	322/2336	303/2124	101/707	V/X	gtC/gt	41317	4318	NM_004994.2	NA	1.04E-04	-
20	44639243	G	A	missense_variant	*MMP9*	36	het	512/2336	493/2124	165/707	D/N	Gac/Aac	42807	4318	NM_004994.2	2.80E-04	4.16E-04	rs8125581
20	44639865	AC	A	frameshift_variant	*MMP9*	NA	het	753/2336	734/2124	245/707	T/X	aCc/ac	41406	4318	NM_004994.2	9.88E-05	2.08E-04	-
20	44639905	C	T	missense_variant	*MMP9*	24.4	het	792/2336	773/2124	258/707	T/I	aCc/aTc	42868	4318	NM_004994.2	9.23E-04	9.36E-04	rs41529445
20	44640295	C	A	stop_gained	*MMP9*	34	het	925/2336	906/2124	302/707	C/*	tgC/tgA	41437	4318	NM_004994.2	1.48E-04	5.20E-04	rs146499495
20	44640295	C	A	stop_gained	*MMP9*	34	het	925/2336	906/2124	302/707	C/*	tgC/tgA	41437	4318	NM_004994.2	1.48E-04	5.20E-04	rs146499495
20	44664420	C	T	missense_variant	*SLC12A5*	23.8	het	429/6041	353/3420	118/1139	P/L	cCa/cTa	42851	57468	NM_001134771.1	1.40E-04	3.12E-04	-
20	44692208	G	C	missense_variant	*NCOA5*	21.5	het	1095/3209	941/1740	314/579	A/G	gCc/gGc	42924	57727	NM_020967.2	8.24E-06	1.04E-04	-
20	44697248	C	T	missense_variant	*NCOA5*	27.1	het	549/3209	395/1740	132/579	R/Q	cGa/cAa	42833	57727	NM_020967.2	8.24E-06	1.04E-04	-
22	38468594	C	T	missense_variant	*PICK1*	29.3	het	959/2109	667/1248	223/415	R/W	Cgg/Tgg	42991	9463	NM_001039583.1	NA	1.04E-04	rs368390981
22	38478793	C	T	missense_variant	*SLC16A8*	36	het	218/1896	88/1515	30/504	G/S	Ggc/Agc	42771	23539	NM_013356.2	4.31E-04	9.36E-04	rs113748161

22	38478793	C	T	missense_variant	*SLC16A8*	36	het	218/1896	88/1515	30/504	G/S	Ggc/Agc	42771	23539	NM_013356.2	4.31E-04	9.36E-04	rs113748161
22	38478793	C	T	missense_variant	*SLC16A8*	36	het	218/1896	88/1515	30/504	G/S	Ggc/Agc	42771	23539	NM_013356.2	4.31E-04	9.36E-04	rs113748161
22	38478793	C	T	missense_variant	*SLC16A8*	36	het	218/1896	88/1515	30/504	G/S	Ggc/Agc	42771	23539	NM_013356.2	4.31E-04	9.36E-04	rs113748161
22	38482409	C	T	missense_variant	*BAIAP2L2*	20.9	het	1451/2145	1307/1590	436/529	R/Q	cGg/cAg	43083	80115	NM_025045.4	3.97E-04	3.12E-04	rs201761880
22	38528920	C	T	missense_variant	*PLA2G6*	20.8	het	1131/3061	995/2259	332/752	C/Y	tGt/tAt	42932	8398	NM_001004426.1	NA	1.04E-04	-
22	38529014	G	A	missense_variant	*PLA2G6*	28.6	het	1037/3061	901/2259	301/752	R/C	Cgc/Tgc	42932	8398	NM_001004426.1	5.97E-05	1.04E-04	rs367854265
22	38541545	G	C	missense_variant	*PLA2G6*	21.4	het	461/3061	325/2259	109/752	H/D	Cac/Gac	42810	8398	NM_001004426.1	4.28E-04	5.20E-04	rs142530390
22	38541545	G	C	missense_variant	*PLA2G6*	21.4	het	461/3061	325/2259	109/752	H/D	Cac/Gac	42810	8398	NM_001004426.1	4.28E-04	5.20E-04	rs142530390