

**Supplementary table 4b. Rare predicted loss of function variants found in controls**

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	allele	Het/hom alt		AminoAcid_positon	AminoAcid_change	CodonChange	ExonNumber	Gene	Transcript	ExAC_AF	Seq_AF	rsid
								cDNA_position	CDS_position									
1	196295941	C	T	missense_variant	KCNT2	27.3	het	2242/5909	2182/3408	728/1135	A/T	Gct/Act	19/28	343450 NM_198503.2	NA	1.04E-04	-	
1	196295991	G	A	missense_variant	KCNT2	23.2	het	2192/5909	2132/3408	711/1135	A/V	gCa/gTa	19/28	343450 NM_198503.2	1.65E-05	2.08E-04	rs377088091	
1	196748330	CTATTTCA	C	frameshift_variant	CFHR3	NA	het	185-191/1457	98-104/810	33-35/289	LFH/X	ctATTTTCAt/ct	41309	10878 NM_001166624.1	3.46E-05	3.12E-04	-	
1	197030944	A	T	missense_variant	F13B	21.7	het	465/2217	421/1986	151/861	S/T	Tct/Act	42806	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	197036552	A	G	splice_donor_variant	F13B	6.958	het	-2472	-1746	-/581	-	-	-	2165 XM_005244955.1	NA	4.16E-04	-	
1	197036552	A	G	splice_donor_variant	F13B	6.958	het	-2472	-1746	-/581	-	-	-	2165 XM_005244955.1	NA	4.16E-04	-	
1	197062180	A	G	splice_donor_variant	ASPM	19.14	het	-16133	-5679	-/1892	-	-	-	259266 NM_001208846.1	1.07E-04	1.04E-04	rs199503603	
1	197062189	C	T	missense_variant	ASPM	32	het	4789/6133	4532/5679	1511/1892	R/Q	cGai/cAa	20/27	259266 NM_001208846.1	3.30E-05	2.08E-04	rs146444278	
1	197062189	C	T	missense_variant	ASPM	32	het	4789/6133	4532/5679	1511/1892	R/Q	cGai/cAa	20/27	259266 NM_001208846.1	3.30E-05	2.08E-04	rs146444278	
2	228173618	C	T	missense_variant	COL4A3	20.3	het	4628/8097	4466/5013	1489/1670	T/I	aCui/aTl	49/52	1285 NM_000091.4	2.48E-05	2.08E-04	rs200818438	
2	228173943	C	T	missense_variant	COL4A3	25.3	het	4826/8097	4664/5013	1555/1670	A/V	gCg/gTg	50/52	1285 NM_000091.4	4.13E-05	1.04E-04	rs369575989	
2	228176554	C	T	missense_variant	COL4A3	23.2	het	5143/8097	4981/5013	1661/1670	R/C	Ccg/Tgc	52/52	1285 NM_000091.4	3.06E-04	6.24E-04	rs201697532	
2	228176554	C	T	missense_variant	COL4A3	23.2	het	5143/8097	4981/5013	1661/1670	R/C	Ccg/Tgc	52/52	1285 NM_000091.4	3.06E-04	6.24E-04	rs201697532	
2	228176554	C	T	missense_variant	COL4A3	23.2	het	5143/8097	4981/5013	1661/1670	R/C	Ccg/Tgc	52/52	1285 NM_000091.4	3.06E-04	6.24E-04	rs201697532	
3	64635321	G	T	missense_variant	ADAMTS9	31	het	1629/7313	1597/5808	533/1935	P/T	Cca/Aca	14885	56999 NM_182920.1	NA	3.12E-04	-	
3	64635321	G	T	missense_variant	ADAMTS9	31	het	1629/7313	1597/5808	533/1935	P/T	Cca/Aca	14885	56999 NM_182920.1	NA	3.12E-04	-	
3	64635449	C	T	missense_variant	ADAMTS9	30	het	1501/7313	1469/5808	490/1935	G/D	gGv/gAl	14885	56999 NM_182920.1	2.31E-04	8.32E-04	rs147650040	
3	99509699	T	C	missense_variant	COL8A1	27	het	432/3091	173/2335	58/744	V/A	gTa/gCa	42830	1295 NM_001850.4	2.97E-04	2.08E-04	-	
4	110603891	T	C	missense_variant	CCDC109B	23.2	het	744/1274	605/1011	202/336	L/P	cTl/cCt	42863	55013 NM_017918.4	3.95E-04	2.08E-04	rs200701059	
4	110603891	T	C	missense_variant	CCDC109B	23.2	het	744/1274	605/1011	202/336	L/P	cTl/cCt	42863	55013 NM_017918.4	3.95E-04	2.08E-04	rs200701059	
4	110605765	C	T	missense_variant	CCDC109B	24	het	918/1274	779/1011	260/336	S/F	tCt/TTT	42894	55013 NM_017918.4	1.65E-05	2.08E-04	-	
4	110605768	G	A	missense_variant	CCDC109B	20.10	het	951/1274	812/1011	271/336	R/Q	cGai/cAa	42894	55013 NM_017918.4	2.22E-04	5.20E-04	rs201083400	
4	110605768	G	A	missense_variant	CCDC109B	20.10	het	951/1274	812/1011	271/336	R/Q	cGai/cAa	42894	55013 NM_017918.4	2.22E-04	5.20E-04	rs201083400	
4	110610551	T	G	missense_variant	CAS9B	32	het	895/1661	817/882	273/293	R/Q	Aag/Cag	42923	839 NM_001226.3	3.30E-05	2.08E-04	rs200347007	
4	110638764	A	G	missense_variant	FLA2G12A	20.8	het	668/5235	391/570	131/189	C/R	Tgc/Tgc	42798	81579 NM_030821.4	5.27E-04	9.36E-04	rs41278045	
4	110638868	C	T	missense_variant	FLA2G12A	33	het	533/5235	256/570	86/189	R/X	Gga/AGA	42770	81579 NM_030821.4	3.30E-05	1.04E-04	-	
4	110650804	G	C	missense_variant	FLA2G12A	34	het	439/5235	162/570	54/189	D/E	gaa/cGg	42770	81579 NM_030821.4	3.30E-05	1.04E-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	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	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	110791550	C	T	stop_gained	LRIT3	38	het	1663/3584	2465/2040	549/679	Q/*	Caa/Taa	41367	345193 NM_198506.4	8.24E-06	1.04E-04	rs377143816	
5	39308855	C	T	stop_gained	C9	37	het	1369/2693	1280/1680	427/559	S/*	tCai/tGa	41527	735 NM_001737.3	8.24E-06	2.08E-04	rs121909594	
5	39389465	A	T	missense_variant	DAB2	26.4	het	1160/4601	629/2313	210/770	V/D	gTl/gAl	42993	1601 NM_001343.3	1.24E-04	4.16E-04	rs140161347	
5	39392564	C	A	missense_variant&splice_region_variant	DAB2	28.2	het	764/4536	233/2250	78/749	V/D	gGg/gTa	42839	1601 NM_00124867.1	NA	1.04E-04	-	
6	31742216	C	T	missense_variant	VARS	21.2	het	3809/4294	3386/3795	1120/1264	R/Q	cGg/cAg	28/30	7407 NM_006295.2	2.97E-04	5.20E-04	rs150145769	
6	31807421	GA	G	frameshift_variant	CBorf48	NA	het	723/842	210/228	70/75	G/X	gg/Agg	41386	50854 NM_001040437.1	NA	3.12E-04	-	
6	31829044	A	G	missense_variant	NEU1	21.4	het	692/2071	536/1248	179/415	V/A	gg/Agg	42800	4758 NM_000434.3	2.54E-05	1.04E-04	rs150302766	
6	31829152	A	T	missense_variant	NEU1	21.8	het	584/2071	428/1248	143/415	V/E	gTg/gAg	42800	4758 NM_000434.3	2.55E-05	1.04E-04	-	
6	31918081	G	A	missense_variant	CFB	20.3	het	1803/2627	1525/2295	509/764	E/K	Cag/Aag	43087	629 NM_001710.5	8.40E-06	2.08E-04	rs144339757	
6	31918081	G	A	missense_variant	CFB	20.3	het	1803/2627	1525/2295	509/764	E/K	Cag/Aag	43087	629 NM_001710.5	8.40E-06	2.08E-04	rs144339757	
6	31918106	CTG	C	frameshift_variant	CFB	NA	het	1829-1830/2627	1551-1552/2295	517-518/764	SE/SX	tCt/Gag/tcag	41625	629 NM_001710.5	NA	1.04E-04	-	
6	31935569	CAG	C	frameshift_variant	SKIV2L	NA	het	3051-3052/4170	2662-2663/3741	888/1248	R/X	AG/g	22/28	6499 NM_069829.4	1.69E-05	1.04E-04	-	
6	31936795	G	C	missense_variant	SKIV2L	26.3	het	3717/4170	3328/3741	1110/1246	A/P	Ccc/Ccc	26/28	6499 NM_069829.4	7.58E-04	2.08E-04	rs200525706	
6	31939288	A	C	missense_variant	MDM3Z	24.1	het	498/1590	165/1191	55/396	D/E	gaT/gaG	42773	1797 NM_005510.3	5.77E-05	2.08E-04	rs199557774	
6	32021216	C	T	missense_variant	TNXB	23.9	het	8936/13125	8734/12729	2912/4242	G/S	Cgc/AGc	25/44	7148 NM_019105.6	6.72E-04	9.36E-04	rs200708257	
6	32021216	C	T	missense_variant	TNXB	23.9	het	8936/13125	8734/12729	2912/4242	G/S	Cgc/AGc	25/44	7148 NM_019105.6	6.72E-04	9.36E-04	rs200708257	
6	32021216	C	T	missense_variant	TNXB	23.9	het	8936/13125	8734/12729	2912/4242	G/S	Cgc/AGc	25/44	7148 NM_019105.6	6.72E-04	9.36E-04	rs200708257	
6	32058615	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	32058615	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	32057030	C	T	missense_variant	TNXB	21.1	het	2687/13125	2485/12729	829/4242	G/S	Cgc/AGc	16193	7148 NM_019105.6	1.34E-03	3.12E-04	rs147172260	
6	32057030	C	T	missense_variant	TNXB	21.1	het	2687/13125	2485/12729	829/4242	G/S	Cgc/AGc	16193	7148 NM_019105.6	1.34E-03	3.12E-04	rs147172260	
6	32057675	C	A	missense_variant	TNXB	21.6	het	413/13125	211/12729	71/4242	V/L	Ctg/Ttg	16103	7148 NM_019105.6	2.57E-04	9.36E-04	rs201922477	
6	32057675	C	A	missense_variant	TNXB	21.6	het	413/13125	211/12729	71/4242	V/L	Ctg/Ttg	16103	7148 NM_019105.6	2.57E-04	9.36E-04	rs201922477	
6	32057675	C	A	missense_variant	TNXB	21.6	het	413/13125	211/12729	71/4242	V/L	Ctg/Ttg	16103	7148 NM_019105.6	2.57E-04	9.36E-04	rs201922477	
6	32057675	C	A	missense_variant	TNXB	21.6	het	413/13125	211/12729	71/4242	V/L	Ctg/Ttg	16103	7148 NM_019105.6	2.57E-04	9.36E-04	rs201922477	
6	32098593	G	A	missense_variant	FKBP1	24.3	het	1235/1344	965/1050	322/349	V/V	gCg/cTc	42768	63943 NM_022110.3	NA	2.08E-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	100084515	G	A	missense_variant		NYAP1	23.5	het	299/3584	140/2526	47/841	R/Q	cGg/cTg	42801	222950	NM_173564.3	2.40E-04	3.12E-04	rs202135611	
7	104747909	G	T	missense_variant		KMT2E	22.5	het	3391/6715	3005/5577	1002/1858	S/I	aGo/aTc	21/26	55904	NM_018682.3	8.24E-06	1.04E-04	-	
7	104748280	G	A	missense_variant		KMT2E	27.4	het	3762/6715	3376/5577	1126/1858	E/K	Gaa/Aaa	21/26	55904	NM_018682.3	5.77E-05	1.04E-04	rs146130548	
8	23082276	AG	A	frameshift_variant		TNFRSF10A	NA	het	404/1750	298/1407	100/468	L/X	Ctg/Tg	41283	8797	NM_003844.3	5.16E-05	1.04E-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	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	23156435	A	G	missense_variant		LXL2	23.7	het	2498/3810	2159/2325	1027/774	V/A	gTt/gTc	13/14	4017	NM_002318.2	5.19E-04	5.20E-04	rs148111808	
8	23167211	C	T	missense_variant		LXL2	20.10	het	2189/3810	1850/2325	617/774	R/H	cGc/cAc	43022	4017	NM_002318.2	4.96E-05	1.04E-04	rs371040157	
8	23174498	C	T	missense_variant		LXL2	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		LXL2	28.5	het	1939/3810	1600/2325	534/774	G/R	Gga/Aga	42992	4017	NM_002318.2	0.013	7.28E-04	rs149587418	
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	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	101832028	C	T	stop_gained		COL15A1	44	het	4376/5422	4027/4167	1343/1388	R/*	Cga/Tga	42/42	1306	NM_001855.4	NA	1.04E-04	-	
9	101980764	C	T	missense_variant		ALG2	34	het	773/2830	703/1251	235/416	E/K	Gaa/Aaa	42768	85365	NM_033087.3	1.65E-05	1.04E-04	rs189167741	
9	101980764	C	T	missense_variant		ALG2	21.6	het	665/2830	595/1251	199/416	L/F	Ctc/Ttc	42768	85365	NM_033087.3	8.24E-06	1.04E-04	-	
9	107558399	C	T	missense_variant		ABCA1	27.6	het	5721/10503	5317/6786	173/2261	V/M	Gtg/Atg	39/50	19	NM_005502.3	1.65E-05	1.04E-04	rs370103164	
9	107558626	T	C	missense_variant		ABCA1	23.7	het	5605/10503	5201/6786	1734/2261	N/S	aAt/aGt	38/50	19	NM_005502.3	NA	2.08E-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/W	gAt/gTg	21/50	19	NM_005502.3	4.37E-04	8.32E-04	rs140365800	
9	107591316	G	A	missense_variant		ABCA1	23	het	2400/10503	1996/6786	666/2261	R/G	Cgg/Tgg	15/50	19	NM_005502.3	4.94E-05	1.04E-04	rs201599169	
9	107592263	G	A	missense_variant&splice_region_variant		ABCA1	21.3	het	1713/10503	1309/6786	437/2261	R/W	Cgg/Tgg	18568	19	NM_005502.3	5.77E-05	1.04E-04	rs150448790	
9	107646756	G	A	missense_variant		ABCA1	32	het	658/10503	254/6786	85/2261	P/L	cCg/cTg	13/14	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	13/14	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	24816911	A	G	missense_variant		KIAA1217	20.5	het	3514/6123	2705/3795	902/1264	Y/C	IAI/IgT	15/19	56243	NM_001098500.1	4.86E-04	5.20E-04	rs138435732	
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	25138812	G	T	stop_gained		PRTFDC1	18.12	het	668/1926	639/678	213/225	C/*	gTt/gTg	41/525	56952	NM_020200.5	1.31E-04	2.08E-04	rs35671637	
10	124187795	C	T	missense_variant		PLEKHA1	34	het	1113/3986	904/1005	302/334	R/W	Cgg/Tgg	13/14	59338	NM_001195608.1	2.39E-04	2.08E-04	-	
10	124187795	C	T	missense_variant		PLEKHA1	34	het	1113/3986	904/1005	302/334	R/W	Cgg/Tgg	13/14	59338	NM_001195608.1	2.39E-04	2.08E-04	-	
10	124189247	CT	C	frameshift_variant		PLEKHA1	NA	het	1258/3881	1009/1215	337/404	F/X	Tt/tTt	41/619	59338	NM_001001974.2	1.89E-04	6.24E-04	-	
10	124248514	G	A	missense_variant		HTRA1	22.9	het	697/2120	569/1443	190/480	R/H	cGc/cAc	41/42	5654	NM_002775.4	4.12E-05	1.04E-04	-	
12	56077768	C	T	stop_gained		METTL7B	18.73	het	879/1506	670/735	224/244	R/*	Cga/Tga	41/306	196410	NM_152637.2	4.00E-03	3.12E-04	rs115687886	
12	56078883	C	T	missense_variant		ITGA7	20.2	het	3605/4136	3385/3426	1129/1141	E/K	Gag/Aag	25/25	3679	NM_001144996.1	1.65E-05	1.04E-04	rs371203166	
12	56078988	G	A	stop_gained		ITGA7	41	het	3500/4136	3280/3426	1094/1141	Q/*	Cag/Tag	25/25	3679	NM_001144996.1	3.05E-04	2.08E-04	rs200390529	
12	56078988	G	A	stop_gained		ITGA7	41	het	3500/4136	3280/3426	1094/1141	Q/*	Cag/Tag	25/25	3679	NM_001144996.1	3.05E-04	2.08E-04	rs200390529	
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	missense_variant		ITGA7	43	het	3801/4502	3448/3489	1150/1162	R/*	Cag/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	missense_variant		ITGA7	43	het	3801/4502	3448/3489	1150/1162	R/*	Cag/Tga	27/27	3679	XM_005268841.1	9.23E-04	8.32E-04	rs139136931	
12	56088642	C	T	missense_variant		ITGA7	25.9	het	2348/4136	2128/3426	710/1141	D/N	Gat/Aat	15/25	3679	NM_001144996.1	8.24E-06	1.04E-04	rs145287267	
12	56088645	C	T	missense_variant		ITGA7	26.6	het	2345/4136	2125/3426	709/1141	G/R	Ggg/Agg	15/25	3679	NM_001144996.1	NA	1.04E-04	-	
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	42/880	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	41/357	3679	NM_001144996.1	1.36E-05	2.08E-04	-	
12	56115149	AC	A	frameshift_variant		RDH5	NA	het	370/1342	182/957	61/318	T/X	aCc/ac	41/309	5959	NM_001199771.1	NA	1.04E-04	rs34619378	
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	56212940	A	C	missense_variant		ORMDL2	23.7	het	249/1334	157/462	53/153	N/H	Aac/Cac	42/770	29095	NM_014182.4	2.80E-03	3.12E-04	rs182538760	
12	56236602	G	A	stop_gained		MMP19	22.2	het	166/3018	13/918	5/305	Q/*	Cag/Tag	4/280	4327	NM_001272101.1	2.67E-03	3.12E-04	rs17844787	
12	112147438	C	T	missense_variant		ACAD10	22	het	840/4107	640/3273	214/1090	L/R	Ctt/Ttt	42/877	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	42/969	80724	NM_001136538.1	7.41E-05	2.08E-04	-	
12	112167654	C	T	stop_gained		ACAD10	42	het	1581/4107	1381/3273	461/1090	R/*	Cga/Tga	41/599	80724	NM_001136538.1	1.81E-04	2.08E-04	rs146335507	
12	112186211	T	C	missense_variant		ACAD10	23.8	het	2869/4107	2669/3273	890/1090	V/A	gTt/gTc	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/gTc	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	112221023	G	A	missense_variant		ALDH2	33.2	het	377/2060	281/1554	94/517	R/H	cGc/cAc	42/807	217	NM_000690.3	6.59E-05	1.04E-04	rs201745983	
12	1122																			



20	44577821	A	T	missense_variant	ZNF335	21.1	het	4012/4452	3889/4029	1297/1342	S/T	Tca/Aca	27/28	63925	NM_022095.3	1.65E-05	1.04E-04	-
20	44578709	C	T	splice_acceptor_variant	ZNF335	14.4	het	-/4452	-/4029	-/1342	-	-	-	63925	NM_022095.3	NA	1.04E-04	-
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	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	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	44592138	G	A	missense_variant	ZNF335	27	het	1630/4452	1507/4029	503/1342	R/C	Cgt/Tgt	43006	63925	NM_022095.3	8.24E-06	1.04E-04	-
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	44592381	G	A	missense_variant	ZNF335	22.6	het	1474/4452	1351/4029	451/1342	R/C	Cgc/Gtc	42975	63925	NM_022095.3	NA	1.04E-04	rs148130868
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	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	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	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	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	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	44641191	G	A	missense_variant	MMP9	27.8	het	1319/2336	1300/2124	434/707	D/N	Gac/Aac	42960	4318	NM_004994.2	1.65E-05	1.04E-04	rs199893610
20	44642364	G	A	stop_gained	MMP9	21	het	1698/2336	1679/2124	560/707	W/*	tGg/tAg	41559	4318	NM_004994.2	9.88E-05	4.16E-04	rs138685704
20	44642364	G	A	stop_gained	MMP9	21	het	1698/2336	1679/2124	560/707	W/*	tGg/tAg	41559	4318	NM_004994.2	9.88E-05	4.16E-04	rs138685704
20	44642364	G	A	stop_gained	MMP9	21	het	1698/2336	1679/2124	560/707	W/*	tGg/tAg	41559	4318	NM_004994.2	9.88E-05	4.16E-04	rs138685704
20	44664420	C	T	missense_variant	SLC12A5	23.8	het	429/6041	353/3420	118/1139	P/L	cCa/cTa	42851	57468	NM_00113477.1	1.40E-04	3.12E-04	-
22	38328647	G	A	missense_variant	MICALL1	35	het	2379/4495	2104/2592	702/863	G/R	Ggg/Agg	43055	85377	NM_033386.3	2.47E-05	2.08E-04	rs368940855
22	38379518	C	G	missense_variant	SOX10	29.9	het	552/2862	274/1401	92/466	V/L	Gtg/Ctg	42770	6663	NM_006941.3	1.98E-04	2.08E-04	rs142113652
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	38508219	A	C	stop_gained	PLA2G6	37	het	2344/3061	2208/2259	736/752	Y/*	taTtaG	16/16	8398	NM_001004426.1	4.95E-05	1.04E-04	rs121908680
22	38528924	C	T	missense_variant	PLA2G6	34	het	1127/3061	991/2259	331/752	D/N	Gac/Aac	42932	8398	NM_001004426.1	NA	1.04E-04	rs199935023
22	38531030	C	T	missense_variant	PLA2G6	27.3	het	995/3061	859/2259	287/752	G/R	Gga/Aga	42902	8398	NM_001004426.1	8.24E-06	1.04E-04	-
22	38536076	C	T	missense_variant	PLA2G6	20.39	het	846/3061	710/2259	237/752	R/H	cGc/cAc	42871	8398	NM_001004426.1	1.24E-04	1.04E-04	rs147066967