Supplementary Table 1: Variants Family 24

genomic (GRCh37/hg19)	Gene	DNA	Protein	Exon	Function	gnomAD	dbSNP	pph2	Sift	CADD	Zygosity	Depth	% Var	gnomAD total	European (non Finnish)	Finnish	Ashkenazi	African	Latino	East Asians	South Asians	Other
10:g.152558036>A	FAM171A1	NM_001010924.1:c.1784C>T	NP_001010924.1:p.Ser595Phe	8/8	missense	251434--4--0		benign	0.04	25.3	het	278	52	251434--4--0	113754--3--0	21612--0--0	10078--0--0	16256--1--0	34592--0--0	18394--0--0	30616--0--0	6132--0--0
10:g.75231333T>A	PPP3CB	NM_001142353.2:c.562A>T	NP_001135825.1:p.Met188Leu	5/14	missense	280032--11--0		benign possibly damaging	0.14	17.49	het	54	56	280032--11--0	128288--5--0	25076--1--0	10300--0--0	24930--0--0	34874--3--0	19798--0--0	29604--0--0	7162--2--0
10:g.84744883G>A	NRG3	NM_001165973.1:c.1022G>A	NP_001159445.1:p.Arg341Gln	11/11	missense	282338--62--0	rs117241349	benign	0.51	10.28	het	40	50	282338--62--0	128952--50--0	25080--5--0	10334--4--0	24924--2--0	35344--0--0	19940--0--0	30572--0--0	7192--1--0
10:g.105127961C>T	TAF5	NM_006951.4:c.215C>T	NP_008882.2:p.Ala72Val	1/11	missense	67512--8--0		benign	0.16	8.322	het	202	50	67512--8--0	29532--6--0	6988--0--0	2230--0--0	9652--0--0	6102--2--0	3778--0--0	7150--0--0	2080--0--0
11:g.124910494C>T	CCDC15	NM_025004.2:c.2743C>T	NP_079280.2:p.Arg915Trp	16/16	missense	271850--28--0		benign	0.01	28	het	199	45	271850--28--0	123908--19--0	24484--0--0	10134--0--0	23434--0--0	34470--1--0	19150--4--0	29276--3--0	6994--1--0
11:g.134119062A>G	**THYN1**	**NM_014174.2:c.479T>C**	**NP_054893.1:p.Met160Thr**	**5/7**	**missense**	**n.l.**		**possibly damaging probably damaging**	**0.02**	**24.6**	**het**	**138**	**46**	**n.l.**								
13:g.39587813T>G	PROSER1	NM_025138.4:c.1576A>C	NP_079414.3:p.Thr526Pro	11/13	missense	n.l.		benign		2.569	het	157	51	n.l.								
13:g.95363524C>T	SOX21	NM_007084.3:c.780G>A	NP_009015.1:p.Met260Ile	1/1	missense	74276--44--0		benign	0.04	22.6	het	78	56	74276--44--0	36236--20--0	6516--22--0	2674--0--0	10084--1--0	6390--1--0	3388--0--0	6862--0--0	2126--0--0
13:g.99446968G>A	DOCK9	XM_005254034.1:c.6194C>T	XP_005254091.1:p.Pro2065Leu	53/53	missense	280528--325--1		benign	0.17 0.16	12.64	het	133	46	280528--325--1	128374--221--1	25020--68--0	10352--0--0	24200--7--0	35342--18--0	35342--1--0	30576--1--0	7130--9--0
15:g.29415549T>G	FAM189A1	NM_015307.1:c.1613A>C	NP_056122.1:p.Gln538Pro	11/11	missense	180766--461--5		probably damaging	0	24.9	het	296	46	180766--461--5	73200--165--1	19916--232--3	7904--2--0	16596--4--0	24310--12--0	12266--0--0	21298--28--1	5276--18--0
15:g.42743265A>G	ZNF106	NM_022473.1:c.1136T>C	NP_071918.1:p.Leu379Pro	2/19	missense	282612--246--0	rs137908783	benign	0.16	7.254	het	148	49	282612--246--0	128958--179--0	25124--2--0	10364--0--0	24968--3--0	35432--49--0	19950--0--0	30610--0--0	7206--13--0
15:g.65345444T>C	SLC51B	NM_178859.3:c.329T>C	NP_849190.2:p.Leu110Ser	4/4	missense	282816--56--0	rs142436732	benign	0.04	21.9	het	94	47	282816--56--0	129154--50--0	25108--0--0	10370--0--0	24958--3--0	35436--3--0	19952--0--0	30616--0--0	7222--0--0
16:g.15120542T>C	PDXDC1	NM_001285445.1:c.1099T>C	NP_001272374.1:p.Phe367Leu	13/22	missense	n.l.		benign	0.72	13.48	het	312	19	n.l.								
16:g.24581399G>T	RBBP6	NM_006910.4:c.3388G>T	NP_008841.2:p.Ala1130Ser	17/18	missense	n.l.		benign	0.73	4.893	het	54	43	n.l.								
16:g.31097725A>G	PRSS53	NM_001039503.2:c.596T>C	NP_001034592.1:p.Leu199Pro	5/11	missense	n.l.		probably damaging	0.18	22.7	het	176	48	n.l.								
16:g.46772043G>A	MYLK3	NM_182493.2:c.581C>T	NP_872299.2:p.Ala194Val	3/13	missense	181988--4--0		benign	0.33	3.659	het	155	37	181988--4--0	86772--1--0	9186--0--0	5246--0--0	12848--0--0	25830--2--0	16698--0--0	20998--1--0	4410--0--0
16:g.55601350A>G	CAPNS2	NM_032330.1:c.682A>G	NP_115706.1:p.Arg228Gly	1/1	missense	248360--2--0		benign	0.17	16.1	het	193	45	248360--2--0	112260--2--0	21534--0--0	10032--0--0	15468--0--0	34466--0--0	17972--0--0	30598--0--0	6030--0--0
16:g.56535282C>T	BBS2	NM_031885.3:c.1208G>A	NP_114091.3:p.Arg403His	10/17	missense	251486--12--0	rs192007013	benign	0.53	19.03	het	74	36	251486--1--0	113764--1--0	21646--0--0	10080--0--0	16256--0--0	34592--0--0	18394--0--0	30616--0--0	6138--0--0
16:g.57789088C>T	KATNB1	NM_005886.2:c.1354C>T	NP_005877.2:p.Pro452Ser	15/20	missense	280286--382--1	rs149569503	benign	0.13	22.3	het	236	46	280286--382--1	127912--312--1	24290--43--0	10300--0--0	24772--11--0	35348--7--0	19908--0--0	30578--0--0	7178--9--0
16:g.67572594C>T	FAM65A	NM_001193523.1:c.184C>T	NP_001180452.1:p.Pro62Ser	3/22	missense	277010--69--0		benign	0.73	18.5	het	222	41	277010--69--0	125302--50--0	25024--12--0	10204--0--0	23850--0--0	35286--1--0	19686--0--0	30556--3--0	7102--3--0
16:g.69390938C>T	TERF2	NM_005652.4:c.1492G>A	NP_005643.2:p.Glu498Lys	10/10	missense	279136--377--0	rs150757154	probably damaging	0.51	34	het	106	45	279136--377--0	128074--276--0	25020--17--0	10204--0--0	24908--13--0	34358--55--0	19778--0--0	29656--2--0	7138--14--0
16:g.69727406A>C	**NFAT5**	**NM_006599.3:c.3624A>C**	**NP_006590.1:p.Leu1208Phe**	**12/14**	**missense**	**n.l.**		**possibly damaging benign**	**0.03**	**12.84**	**het**	**109**	**47**	**n.l.**								
16:g.72821618_72821624delCCGCCG	ZFHX3	NM_006885.3:c.10551_10556del	NP_008816.3:p.Gly3526_Gly3527del	10/10	indel	139356--78--2	rs112443847				het	229	50	139356--78--2	71090--62--1	14858--4--0	5306--4--0	13534--0--0	12170--1--0	6668--4--1	12466--1--0	3264--2--0
16:g.75301868G>C	BCAR1	NM_001170715.1:c.11C>G	NP_001164186.1:p.Pro4Arg	1/7	missense	248506--3--0		benign	0	10.36	het	52	50	248506--3--0	112516--2--0	20826--0--0	9276--0--0	21646--0--0	32624--0--0	18372--0--0	26784--1--0	6462--0--0
16:g.82101883G>A	HSD17B2	NM_002153.2:c.374G>A	NP_002144.1:p.Arg125Gln	2/5	missense	251432--3--0		benign	0.22	22.5	het	124	51	251432--3--0	113740--1--0	21646--0--0	10078--0--0	16256--0--0	34584--1--0	18382--0--0	30610--1--0	6136--0--0
17:g.3301171G>C	OR1E1	NM_003553.2:c.534C>G	NP_003544.2:p.Phe178Leu	1/1	missense	n.l.		possibly damaging	0	26.5	het	237	46	n.l.								
18:g.57020508T>C	LMAN1	NM_005570.3:c.565A>G	NP_005561.1:p.Ser189Gly	5/13	missense	n.l.		benign	1	8.51	het	113	48	n.l.								
1:g.12939694T>A	PRAMEF4	NM_001009611.4:c.1108A>T	NP_001009611.2:p.Asn370Tyr	4/4	missense	276654--6--0	rs201682396	possibly damaging	0.12	19.3	het	86	35	276654--6--0	126322--5--0	24772--1--0	10182--0--0	23878--0--0	34324--0--0	19778--0--0	30356--0--0	7042--0--0
1:g.210536249G>A	HHAT	NM_001170564.2:c.145G>A	NP_001164035.1:p.Gly49Arg	3/10	missense	282792--168--0	rs141591165	benign	0.69	15.29	het	110	46	282792--168--0	129146--158--0	25112--7--0	10368--0--0	24954--4--0	35426--7--0	19952--0--0	30614--6--0	7220-4--0
7:g.102448760C>T	FAM185A	NM_001145269.1:c.739C>T	NP_001138741.1:p.Arg247Cys	7/7	missense	188864--108--0	rs145625209	benign possibly damaging	0.05	15.86	het	72	44	188864--108--0	76600--74--0	20366--1--0	8802--23--0	16824--2--5	25518--5--0	12458--0--0	22772--1--0	5524--2--0
7:g.129107275A>C	STRIP2	NM_020704.2:c.1861A>C	NP_065755.1:p.Ile621Leu	17/21	missense	250768--1--0	rs745785530	benign probably damaging	0.09	31	het	102	57	250768--1--0	113528--1--0	21356--0--0	10070--0--0	16232--0--0	34498--0--0	18390--0--0	30582--0--0	6112--0--0
1:g.2520973C>T	FAM213B	ENST00000378427.1:c.*120C>T		7/7	3_prime_UTR_variant		rs111819834			5.094	het	304	50	31354--40--0	15404--15--0	3474--2--0	290--0--0	8694--16--0	848--1--0	1560--4--0	0--0--0	1084--2--0
10:g.44102498G>A	ZNF485	NM_001318140.1:c.-1G>A		2/5	5_prime_UTR_variant		rs201767643			16.27	het	144	49	31354--40--0	15404--15--0	3474--2--0	290--0--0	8694--16--0	848--1--0	1560--4--0	0--0--0	1084--2--0
16:g.67472349*82del	ATP6V0D1	NM_004691.5:c.*53_*82del		8/8	3_prime_UTR_variant		rs1265758032				het	52	33	31308--1--0	15388--1--0	3472--0--0	288--0--0	8666--0--0	848--0--0	1558--0--0	0--0--0	1088--0--0
18:g.12277434A>G	CIDEA	NM_001279.4:c.*165A>G		5/5	3_prime_UTR_variant		rs1461929091			0.849	het	27	48	30426--1--0	14976--1--0	3264--0--0	284--0--0	8526--0--0	786--0--0	1544--0--0	0--0--0	1046--0--0
5:g.435041G>A	AHRR	NM_001242412.1:c.*92G>A		11/11	3_prime_UTR_variant		rs557100813			0.312	het	35	43	31408--20--0	15432--14--0	3474--0--0	290--1--0	8716--1--0	848--1--0	1560--0--0	0--0--0	1088--3--0
7:g.94285513G>A	SGCE	NM_001346719.2			5utr regulation		rs1404487860			13.88	het	178	48	n.l.								
7:g.138279297G>A	SVOPL	NM_001139456.1:c.*58G>A		15/15	3_prime_UTR_variant		rs565695569			3.922	het	65	52	31336--16--0	15412--12--0	3448--0--0	290--0--0	8702--3--0	842--1--0	1558--0--0	0--0--0	1084--0--0
13:g.77900914-4del	MYCBP2	NM_015057.5:c.-15_-4del		1/83	5_prime_UTR_variant		rs779632297				het	33	45	30496--20--0	14948--0--0	3364--0--0	282--0--0	8450--20--0	826--0--0	1556--0--0	0--0--0	1070--0--0
16:g.46598149G>C	ANKRD26P1	NR_026556.1:n.496G>C		2/17	transcribed_pseudogene					3.531	het	120	43	n.l.								
17:g.74865050G>T	MGAT5B	NM_001199172.2:c.-62G>T		1/18	5_prime_UTR_variant					3.357	het	85	49	n.l.								

gnomAD: Number of alleles--heterozygote carriers--homozygote carriers, n.l. not listed (https://gnomad.broadinstitute.org/)
pph2
CADD: Combined Annotation Dependent Depletion score (https://cadd.gs.washington.edu/)
Sift:
Zygosity: het: heterozygous, hom: homozygous
Depth: read depth of the variant at the position
%Var: precentage of reads of the alternate variant
n.l.: not listed
bold: all gene variants discussed in the manuscript

Supplementary Table 1: Variants Family 21

genomic (GRCh37/hg19)	Gene	DNA	Protein	Exon	Function	gnomAD	dbSNP	pph2	Sift	CADD	Zygosity	Depth	% Var	gnomAD total	European (non Finnish)	Finnish	Ashkenazi	African	Latino	East Asians	South Asians	Other
18:g.74700385A>T	**MBP**	**NM_001025101.1:c.750+66T>A**			**intron_variant**	**n.l.**	**n.a.**	**benign**	**0.02**	**0.11**	**het**	**200**	**45**	n.l.								
		uc010xfe.1: c.417T>A	B7Z3Y6-1:p.Phe139Leu	4/4	missense																	
4:g.143226886C>A	**INPP4B**	**NM_003866.3:c.424-196G>T**			**intron_variant**	**1/95788**		**benign**	**0**	**7.8**	**het**	**60**	**45**	95844--1--0	40850--1--0	5256--0--0	5694--0--0	5630--0--0	12314--0--0	8198--0--0	15018--0--0	2884--0--0
		uc011chp.1:c.3G>T	XP_005263380.1:p.Met1?	1/13	start lost																	
1:g.158263345G>A	CD1C	NM_001765.3:c.*79G>A		6/6	3_prime_UTR_variant		rs374456855			2.509	het	92	46	282274--120--0	128852--99--0	25106--1--0	10360--0--0	24884--10--0	35376--6--0	19910--0--0	30588--1--0	7198--3--0
6:g.139094704C>T	CCDC28A	NM_015439.2:c.-108C>T		1/6	5_prime_UTR_variant		rs778077003			11.29	het	260	45	238864--1--0	108926--1--0	16622--0--0	9914--0--0	14516--0--0	34322--0--0	18060--0--0	30568--0--0	5936--0--0
7:g.142630638A>G	TRPV5	NM_019841.7:c.-82A>G		1/15	5_prime_UTR_variant		rs139060958			4.394	het	186	50	31384--60--1	15428--2--0	3476--0--0	290--0--0	8700--7--0	846--26--1	846--18--0	0--0--0	1088--7--0
chrX:149681118C>T	MAMLD1	NM_001177466.2:c.*889C>T		6/6	3_prime_UTR_variant		rs781858622			n.a.	het	260	49	121584--240--92	6695--115--49	50993--115--39	6282--0--0	10962--2--1	10962--0--0	8815--0--0	14029--0--0	3891--8--3

Supplementary Table 1: Variants Family 13

genomic (GRCh37/hg19)	Gene	DNA	Protein	Exon	Function	gnomAD	dbSNP	pph2	Sift	CADD	Zygosity	Depth	% Var	gnomAD total	European (non Finnish)	Finnish	Ashkenazi	African	Latino	East Asians	South Asians	Other
3:g.16306352T>G	DPH3	NM_206831.2:c.32A>C	NP_996662.1:p.Glu11Ala	1/3	missense_variant	n.l.		probably damaging possibly damaging	0.02	29.5	het	190	48	n.l.								

Supplementary Table 1: Variants Family 12

genomic (GRCh37/hg19)	Gene	DNA	Protein	Exon	Function	dbSNP	pph2	Sift	CADD	Zygosity	Depth	% Var	gnomAD total	European (non Finnish)	Finnish	Ashkenazi	African	Latino	East Asians	South Asians	Other
10:g.123923476A>C	TACC2	NM_006997.2	NP_008928.1:	1/17	missense intronic		benign	0.12	16.9	het	139	45	237670–29–0	104948–29–0	21196–0–0	9432–0–0	20714–0–0	32276–0–0	17086–0–0	25724–0–0	6294–0–0
12:g.48105483T>C	ENDOU	NM_001172423	NP_001165913	9/10	missense intronic	rs112570968	benign possibly damaging	0.81	23.7	het	75	45	282744–71–0	129110–50–0	25096–0–0	10368–0–0	24960–18–0	35424–3–0	19950–0–0	30612–0–0	7224–0–0
12:g.49993357C>T	FAM186B	NM_032130.2	NP_115506.1:	4/7	missense		benign		6.889	het	165	50	280936–53–0	128104–8–0	25018–0–0	10228–0–0	24948–1–0	35184–2–0	19900–0–0	30376–42–0	7178–0–0
12:g.65448898T>C	WIF1	NM_007191.4	NP_009122.2:	9/10	missense		probably damaging		24.4	het	92	43	250560–1–0	113490–1–0	21636–0–0	10042–0–0	16232–0–0	34382–0–0	18324–0–0	30348–0–0	6106–0–0
13:g.35734035G>C	NBEA	NM_015678.4	NP_056493.3:	22/58	missense		benign	0.09	16.81	het	178	46	223174–1–0	98830–1–0	20418–0–0	9444–0–0	13524–0–0	31664–0–0	16084–0–0	27642–0–0	5568–0–0
13:g.96515937T>A	UGGT2	NM_020121.3	NP_064506.3:	31/39	missense	rs149785094	possibly damaging	0	29.8	het	51	35	282360–120–1	128838–104–1	25096–0–0	10366–2–0	24950–1–0	35358–7–0	19948–0–0	30600–0–0	7204–6–0
14:g.21961355A>G	TOX4	NM_014828.2	NP_055643.1:	7/9	missense		benign	0.35	7.785	het	153	50	282418–27–0	128810–26–0	25112–0–0	10356–0–0	24944–0–0	35420–0–0	19954–0–0	30616–1–0	7206–0–0
14:g.22102976G>A	OR10G2	NM_001005548	NP_001005461	1/1	missense		probably damaging	0.18	22.1	het	174	44	276768–32–0	126198–29–0	24862–0–0	9884–0–0	24548–0–0	34768–1–0	19836–0–0	29634–1–0	7038–1–0
14:g.23745014G>C	HOMEZ	NM_020834.2	NP_065885.2:	2/2	missense		possibly damaging	0.27	7.663	het	272	53	277014–354–1	126736–251–1	24748–3–0	9922–20–0	24160–7–0	35136–58–0	19530–0–0	29738–0–0	7044–15–0
15:g.25940059G>A	ATP10A	NM_024490.3	NP_077816.1:	14/21	missense	rs147432497	probably damaging	0.06	26.4	het	184	39	282532–237–0	128878–161–0	25118–53–0	10358–0–0	24960–0–0	35430–0–0	19954–17–0	30616–0–0	7218–6–0
15:g.31775847G>A	OTUD7A	NM_130901.2	NP_570971.1:	14/14	missense		benign	0.09	22.5	het	42	36	n.l.								
15:g.52901454T>C	FAM214A	NM_019600.3	NP_062546.2:	6/13	missense noncoding		benign	0.29	0.001	het	51	39	n.l.								
16:g.58073910G>A	MMP15	NM_002428.3	NP_002419.1:	4/10	missense		possibly damaging	0.38	22.5	het	216	45	251140–1–0	21618–0–0	113520–0–0	10070–0–0	16224–0–0	34578–0–0	18384–1–0	30616–0–0	6130–0–0
16:g.75563835C>T	CHST5	NM_024533.4	NP_078809.2:	3/3	missense	rs148051752	probably damaging	0.19	27.5	het	445	45	280686–361–1	127620–317–1	25106–23–0	10316–2–0	24598–11–0	35378–5–0	19880–0–0	30592–1–0	7196–2–0
16:g.89620912G>T	SPG7	NM_003119.3	NP_003110.1:	16/17	missense		benign	0.68	20.8	het	120	58	n.l.								
18:g.74620491G>A	ZNF236	NM_001306082	NP_001293011	14/31	missense noncoding		benign	0.57	8.494	het	91	56	279092–3–0	128054–3–0	23974–0–0	10332–0–0	24166–0–0	35346–0–0	19522–0–0	30570–0–0	7128–0–0
19:g.308813G>A	MIER2	NM_017550.2	NP_060020.1:	11/14	missense		benign	0.05	25.2	het	137	55	280804–179–1	127870–141–0	24994–27–1	10270–0–0	24796–3–0	35344–3–0	19782–1–0	30572–0–0	7176–4–0
19:g.548979C>T	GZMM	NM_001258335	NP_001245288	4/5	missense	rs140428817	possibly damaging	0.06	22.3	het	40	65	263458–17–0	118060–4–0	23410–0–0	9704–0–0	22982–7–0	34350–1–0	19100–1–0	29120–4–0	6732–0–0
19:g.1506821C>A	ADAMTSL5	NM_213604.2	NP_998769.2:	10/12	missense		possibly damaging	0.19	23.4	het	61	56	173280–68–0	69918–41–0	18606–2–0	7970–10–0	16202–1–0	21712–2–0	12076–0–0	21724–10–0	5072–2–0
19:g.2115611C>T	AP3D1	NM_001261818	NP_001248751	19/32	missense		benign	0.6	19.4	het	129	57	247612–1–0	112122–1–0	21238–0–0	10000–0–0	15342–0–0	34402–0–0	17922–0–0	30568–0–0	6018–0–0
19:g.9579809C>A	ZNF560	NM_152476.2	NP_689689.2:	9/10	missense	rs61737643	benign	0.18	17.21	het	45	49	257102–503–1	118558–446–1	24374–9–0	9574–0–0	23774–17–0	29826–16–0	18682–0–0	25664–0–0	6650–15–0
19:g.11363543A>G	DOCK6	NM_020812.3	NP_065863.2:	3/48	missense		possibly damaging	0	21.3	het	104	40	248566–3–0	112518–0–0	21538–0–0	10040–0–0	15408–0–0	34458–3–0	17968–0–0	30594–0–0	6042–0–0
1:g.1222333G>A	SCNN1D	NM_001130410	NP_001123888	8/18	missense 5utr	rs145306516	benign	0.79	7.488	het	122	47	278760–105–0	126446–73–0	24948–8–0	10184–0–0	24436–10–0	35302–7–0	19772–0–0	30540–4–0	7132–3–0
1:g.2938266G>A	ACTRT2	NM_080431.4	NP_536356.3:	1/1	missense	rs141283101	benign	0	0.056	het	63	38	281666–53–0	128492–24–0	25088–1–0	10276–0–0	24742–4–0	35366–15–0	19940–0–0	30578–7–0	7184–2–0
1:g.3413612G>C	MEGF6	NM_001409.3	NP_001400.3:	28/37	missense intronic		benign	0.48	10.04	het	90	48	188962–209–0	76526–167–0	18134–1–0	8740–1–0	17100–9–0	26358–18–0	13292–0–0	23332–7–0	5480–6–0
1:g.12785721G>A	AADACL3	NM_001103117	NP_00109664	4/4	missense	rs185949240	possibly damaging	0.02	22.5	het	125	45	280702–132–0	128466–119–0	25036–5–0	10352–0–0	24198–3–0	35372–1–0	19534–0–0	30602–3–0	7142–1–0
1:g.17273371C>T	CROCC	NM_014675.3	NP_055490.3:	17/37	missense	rs41267493	benign	0.13	25.6	het	97	39	197826–556–0	81512–196–0	19602–2–0	8862–157–0	18416–6–0	26380–24–0	14102–0–0	23260–158–0	5692–13–0
1:g.18703695A>G	CLCA4	NM_012128.3	NP_036260.2:	12/14	missense		benign	0.1	8.009	het	132	44	247420–3–0	111856–0–0	21444–0–0	10018–0–0	15412–2–0	34304–0–0	17884–0–0	30522–1–0	5980–0–0
22:g.22040839A>G	PPIL2	NM_148175.2	NP_680480.1:	11/21	missense		benign	0.28	21.3	het	165	45	n.l.								
22:g.25255728A>G	SGSM1	NM_001039994	NP_00103503	9/26	missense		benign	0.22	18.53	het	219	49	n.l.								
22:g.31971368C>G	SFI1	NM_001007462	NP_00100746	10/33	missense noncoding		probably damaging possibly damaging	0.07	23.1	het	29	55	n.l.								
22:g.50648628C>T	SELO	NM_031454.1	NP_113642.1:	4/9	missense 5utr	rs543629121	possibly damaging	0.05	23.9	het	62	44	247564–17–0	111874–4–0	21442–0–0	9990–0–0	15274–0–0	34446–5–0	17934–2–0	30590–4–0	6014–2–0
2:g.28719012G>C	PLB1	NM_001170582	NP_001164053	1/57	missense		benign	0.56	5.662	het	99	38	251096–4–0	113434–4–0	21626–0–0	10064–0–0	16256–0–0	34588–0–0	18394–0–0	30612–0–0	6122–0–0
2:g.44055163C>T	ABCG5	XM_005264487	XP_005264543	5/12	missense 5utr	rs141828689	probably damaging	0.02	34	het	141	55	282590–370–0	128988–290–0	25076–45–0	10368–0–0	24956–3–0	35426–20–0	19950–0–0	30604–1–0	7222–1–0
2:g.109237865C>T	LIMS1	NM_001193487	NP_001180411	1/10	nonsense intronic regulation	rs56186549		0.89	35	het	190	44	182604–232–0	71648–189–0	20484–8–0	8708–0–0	15878–5–0	25482–19–0	12356–1–0	22536–2–0	5512–8–0
2:g.121991702T>C	TFCP2L1	NM_014553.2	NP_055368.1:	12/15	missense noncoding	rs139334996	benign	0.56	16.45	het	32	44	282858–655–1	129196–419–1	25094–139–0	10368–10–0	24970–12–0	35438–29–0	19952–0–0	30614–29–0	7226–17–0
2:g.175201171G>A	SP9	NM_001145252	NP_001138722	2/2	missense		benign	0.41	3.977	het	110	42	66276–3–0	23948–2–0	5680–0–0	4314–0–0	896–0–0	12604–0–0	3404–0–0	13406–1–0	2024–0–0
2:g.202508123T>C	TMEM237	NM_001044382	NP_00103785	1/13	missense 5utr regulation / Start Lost		possibly damaging	0.03	13.94	het	65	51	126266–5–0	45262–3–0	14482–0–0	7690–0–0	4228–0–0	22270–2–0	7728–0–0	21018–0–0	3588–0–0
3:g.38674658G>A	SCN5A	NM_001099404	NP_00109928	7/28	syn missense	rs571894716			7.846	het	63	52	278234–5–0	126958–1–0	24846–0–0	10288–0–0	23982–0–0	35198–4–0	19396–0–0	30474–0–0	7092–0–0
3:g.39156160C>T	**TTC21A**	**NM_145755.2**	**NP_665698.2:**	**6/29**	**missense noncoding**		**probably damaging**	**0.15**	**0.26**	**26.6**	**het**	**102**	**52**	n.l.							
3:g.49723695G>A	MST1	NM_020998 (u	Arg355Trp	8/10	missense					6.417	het	26	58	n.l.							
3:g.69244405T>C	FRMD4B	NM_015123.2	NP_055938.1:	15/23	missense intronic		probably damaging possibly damaging	0.12	26.7	het	193	53	279712–103–0	128154–81–0	24982–2–0	10304–1–0	24184–3–0	35108–14–0	19460–0–0	30420–1–0	7100–1–0
3:g.100645259C>T	ABI3BP	NM_015429.3	NP_056244.2:	2/35	missense		probably damaging possibly damaging	0.03	34	het	280	46	280306–110–2	128250–32–1	25020–4–0	10344–1–0	24182–1–0	35278–2–0	19506–0–0	30594–64–1	7132–6–0
4:g.89397119G>A	HERC5	NM_016323.3	NP_057407.2:	12/23	missense		benign	0.3	8.884	het	209	47	n.l.								
4:g.103968215A>C	SLC9B2	NM_178833.5	NP_849155.2:	7/12	missense		probably damaging possibly damaging	0.29	26.8	het	107	57	250760–3–0	113290–3–0	21620–0–0	10044–0–0	16240–0–0	34474–0–0	18386–0–0	30598–0–0	6108–0–0
5:g.137213294G>A	MYOT	NM_001300911	NP_00128784	5/11	missense	rs151094883	benign	0.04	0.166	het	174	53	282852–185–0	129164–20–0	25120–0–0	10368–0–0	24968–1–0	35436–1–0	19954–142–0	30616–16–0	7226–5–0
6:g.6175022A>T	F13A1	NM_000129.3	NP_000120.2:	12/15	missense		benign	0.47	0.008	het	119	49	n.l.								
6:g.109330635G>A	SESN1	NM_001199952	NP_00118686	1/10	missense intronic regulation	rs142983751	benign	0.1	10.54	het	36	47	87234–117–1	36916–54–0	8212–0–0	5390–1–0	9306–2–0	9088–1–0	2818–0–0	12930–48–1	2574–1–0
6:g.144835942A>C	UTRN	NM_007124.2	NP_009055.2:	36/74	missense	rs144388800	probably damaging	0.53	32	het	156	44	280598–433–0	128482–251–0	25084–84–0	10240–3–0	24948–12–0	34864–48–0	19846–0–0	29962–25–0	7172–10–0
7:g.128547443G>A	KCP	NM_001135591	NP_00112938	4/38	nonsense				11.08	het	54	41	n.l.								
7:g.149544983T>C	ZNF862	NM_001099222	NP_00109269	4/8	missense		benign	0.27	0.588	het	227	53	**n.l.**								
7:g.154429580A>G	DPP6	NM_001039935	NP_00103443	6/26	missense intronic		benign	0.1	16.9	het	139	45	191484–1–0	80434–1–0	18420–0–0	9084–0–0	12412–0–0	27240–0–0	14264–0–0	24534–0–0	5096–0–0
9:g.85627390C>A	RASEF	NM_152573.3	NP_689786.2:	5/17	missense	rs140992010	possibly damaging	0	32	het	71	45	258286–194–0	119040–1–0	24390–0–0	9692–0–0	23170–181–0	31120–8–0	17726–0–0	26536–0–0	6612–4–0
9:g.96209957T>C	FAM120AOS	NM_001322222	NP_00130915	3/3	missense noncoding	rs113476650	benign	0	3.251	het	107	53	282622–722–1	129060–446–1	25108–35–0	10366–128–0	24962–15–0	35374–54–0	19946–0–0	30594–20–0	7212–24–0
9:g.114994395G>C	PTBP3	NM_001163755	NP_001157265	10/15	missense intronic	rs144684166	benign	0.09	22.7	het	148	52	280866–66–0	128574–51–0	25064–6–0	10316–0–0	24918–0–0	34872–6–0	19810–0–0	30146–0–0	7166–3–0
9:g.121930417G>A	BRINP1	NM_014618.2	NP_055433.2:	8/8	missense	rs149284594	possibly damaging	0	26.1	het	89	48	267540–102–0	121782–71–0	24054–11–0	9404–0–0	23784–4–0	33804–5–0	19496–2–0	28366–7–0	6850–2–0
9:g.139360491C>T	SEC16A	NM_014866.1	NP_055681.1:	8/32	missense		benign possibly damaging	0.45	23.1	het	112	40	248714–3–0	112654–2–0	21484–0–0	10034–0–0	15446–0–0	34496–0–0	17964–1–0	30596–0–0	6040–0–0
9:g.139371628C>A	SEC16A	NM_014866.1	NP_055681.1:	3/32	missense		benign		23.4	het	149	51	n.l.								
9:g.140605429C>T	EHMT1	NM_024757.4	NP_079033.4:	2/27	missense intronic		benign	0.02	25.2	het	247	49	251128-24–0	113906–21–0	22508–0–0	9708–0–0	21296–1–0	32468–1–0	17880–0–0	26810–1–0	6552–0–0
X:g.30236743C>T	MAGEB2	NM_002364.4	NP_002355.2:	2/2	missense		probably damaging	0.13		het	112	46	202545– 18–0 (6Hemiz	91462–10–0	18548–0–0	7514–0–0	18924–1–0	27688–1–0	14685–0–0	18461–6–0	5263–0–0
X:g.44929178T>A	KDM6A	NM_001291417	NP_00127834	17/29	missense noncoding		benign	1		het	293	44	n.l.								
X:g.83128179C>G	CYLC1	NM_021118.2	NP_066941.1:	4/5	missense intronic		benign	1		het	75	45	194607–94–1 (28Hemi	87707–31–0	18309–6–0	7243–0–0	18061–1–0	26816–36–1	14288–0–0	17132–3–0	5051–17–0
X:g.150912051T>C	CNGA2	NM_005140.1	NP_005131.1:	7/7	missense		benign	0.05		het	131	41	205174–7–0 (2Hemizy	92694–6–0	18537–0–0	7665–0–0	18973–0–0	28048–1–0	14846–0–0	19076–0–0	5335–0–0
13:20797308-20797309 C	GJB6	NM_001110219	NP_00110368	5/5	frameshift					het	169	53	251254–6–0	113644–5–0	21614–0–0	10064–0–0	16246–0–0	34566–0–0	18394–0–0	30600–0–0	6126–1–0
18:48723148-48723154	MEX3C	NM_016626.4:	c.537_542del	1/2	inframe_deletion					het	88	39	149532–12–0	60826–2–0	9742–0–0	7602–0–0	12812–0–0	22842–0–0	9882–10–0	21074–0–0	4752–0–0
3:40457449-40457450	ENTPD3	NM_001291969	NP_00127888	7/11	frameshift					het	159	48	n.l.								
3:97499054-97499055	ARL6	NM_001323515	NP_00131044	3/9	frameshift					het	50	56	n.l.								
10:g.104849385C>A	NT5C2	NM_001351169.1:	c.*44C>A	19/19	3_prime_UTR_variant	rs191195125			11.65	het	15	47	216556–9–0	100670–1–0	21492–0–0	5200–0–0	23108–0–0	26342–0–0	17520–8–0	16744–0–0	5480–0–0
11:g.429534C>T	ANO9	NM_001012302.3:	c.915+36C>T		5utr intronic	rs202000265			0.412	het	151	41	273314–26–0	123754–1–0	23304–0–0	10120–0–0	24178–0–0	35054–1–0	19614–22–0	30292–2–0	6998–0–0
11:g.6584485G>A	DNHD1	NM_144666.2:	c.9669+124G>A		intronic variant	rs184807755			0.91	het	167	49	180806–48–0	71814–37–0	18382–0–0	8644–1–0	16012–0–0	25444–7–0	12430–0–0	22730–0–0	5350–3–0
14:g.72205924C>T	SIPA1L1	NM_001284245.3		26/26	3_prime_UTR_variant				1.069	het	62	44	251814–456–0	117208–381–0	22706–117–0	7276–21–0	24434–5–0	30258–11–0	19146–5–0	24524–5–0	6262–11–0
17:g.3912331G>A	ZZEF1	NM_015113.4:	c.8734-83G>A		intronic variant	rs753847981			2.624	het	12	50	31390–4–0	15426–3–0	3472–0–0	290–0–0	8712–1–0	848–0–0	1556–0–0	0–0–0	1086–0–0
19:g.376010C>T	THEG	NM_016585.5:	c.-40C>T	1/8	5_prime_UTR_variant	rs200584728			0.909	het	14	56	137348-148-2	73586–126–2	9686–20–0	2168–0–0	17868–0–0	12464–0–0	9956–1–0	8402–0–0	3218–1–0
19:g.48182604C>T	GLTSCR1	NM_015711.3	NP_056526.3:	6/15	synonymous_variant	rs1052849514			6.9	het	117	46	59746–0–0	24890–0–0	11990–0–0	1136–0–0	3642–0–0	7352–0–0	5058–0–0	4002–0–0	1676–0–0
2:g.27579006T>C	GTF3C2	NM_001318909.1:	c.-44T>C	1/19	5_prime_UTR_variant	rs777495786			11.4	het	216	53	166624–48–0	68744–10–0	8864–1–0	8608–0–0	15232–0–0	25322–3–0	12108–0–0	22494–33–0	5252–1–0
2:g.74687826G>A	INO80B	NM_031288.4			downstream_gene_variant	rs370974475			0.876	het	30	53	213504–34–0	101530–32–0	20664–0–0	4226–0–0	23472–0–0	24426–2–0	18018–0–0	15876–0–0	5292–0–0
2:g.119752106C>G	MARCO	NM_006770.3:	c.*10C>G	17/17	3_prime_UTR_variant	rs111933917			4.315	het	47	45	240954–53–0	113452–0–0	22112–0–0	6544–0–0	24360–49–0	28604–4–0	18760–0–0	21156–0–0	5966–0–0
2:g.202153341A>T	ALS2CR12	NM_001127391.3:	c.*37A>T	15/15	3_prime_UTR_variant	rs1396954699			6.154	het	146	54	247420–1–0	112144–1–0	21440–0–0	9836–0–0	16194–0–0	33488–0–0	18284–0–0	30010–0–0	6024–0–0
2:g.204131293C>G	CYP20A1	NM_001371702.1:	c.-117C>G	4/11	5_prime_UTR_variant	rs148045812			13.84	het	201	47	282856–17–0	129166–9–0	25120–0–0	10370–0–0	24968–0–0	35440–0–0	19950–0–0	30616–8–0	7226–0–0
2:g.216003041G>A	ABCA12	NM_173076.3:	c.-110G>A	1/53	5_prime_UTR_variant	rs754004029			9.716	het	124	49	31336–1–0	15382–1–0	3468–0–0	290–0–0	8702–0–0	848–0–0	1558–0–0	0–0–0	1088–0–0
3:g.49851284C>T	UBA7	NM_003335.3:	c.-64C>T	1/24	5_prime_UTR_variant	rs189563401			7.814	het	138	51	31394–35–0	15418–21–0	3472–0–0	290–8–0	8718–4–0	848–1–0	1560–0–0	0–0–0	1088–1–0
3:g.180707419G>T	DNAJC19	NM_145261.4:	c.-29G>T	1/6	5_prime_UTR_variant	rs181271620			11.45	het	158	45	227874–282–0	99802–222–0	20370–15–0	9206–1–0	20250–8–0	30260–2–0	16458–0–0	25336–29–0	6192–5–0
7:g.100779012C>T	SERPINE1	NM_000602.5	NP_000593.1:	7/9	syn 3utr regulation	rs756727666			12.84	het	118	42	251424–5–0	113730–1–0	21620–0–0	10080–0–0	16256–0–0	34592–0–0	18392–2–0	30614–2–0	6140–0–0
7:g.120969765G>A	WNT16	NM_016087.2	NP_057171.2:	2/4	syn 5utr	rs1210772767			12.74	het	110	44	250332–1–0	113360–1–0	21092–0–0	10062–0–0	16168–0–0	34578–0–0	18352–0–0	30610–0–0	6110–0–0
9:g.35072351C>T	VCP	NM_001354927.1:	c.-196C>T	1/17	5_prime_UTR_variant	rs1462477819			19.22	het	52	48	82488–0–0	30656–0–0	4790–0–0	6380–0–0	972–0–0	16136–0–0	4034–0–0	17080–0–0	2440–0–0
9:g.112898487T>C	PALM2-AKAP2	NM_001136562.3:	c.-31T>C	2/4	5_prime_UTR_variant	rs141992704			0.139	het	174	45	280270–41–0	126700–32–0	25102–0–0	10342–1–0	24968–1–0	35416–4–0	19954–0–0	30586–0–0	7202–3–0
22:g.19710735C>A	SEPT5	NM_001009939.3:	c.*813C>A	11/11	3_prime_UTR_variant	rs562942429			9.871	het	46	41	161694–240–0	64980–126–0	8864–73–0	8388–16–0	14788–3–0	25204–15–0	11992–0–0	22384–0–0	5094–7–0
9:g.124751792A>G	TTLL11	NM_194252.2	NP_919228.2:	4/4	syn 3utr noncoding	rs115487738			6.898	het	125	52	282872–229–2	129192–98–1	25114–0–0	10370–0–0	24966–1–0	35438–14–0	19950–6–0	30616–96–1	7226–14–0
9:g.139306503C>T	PMPCA	NM_001282946.1:	c.-173C>T	2/13	syn 5utr noncoding regulation	rs142694992			14.06	het	111	47	282752–290–0	129094–219–0	25122–10–0	10364–1–0	24948–6–0	35432–28–0	19952–0–0	30616–18–0	7224–8–0
X:g.106308898C>T	RBM41	NM_001324244.2:	c.*86C>T	9/9	3_prime_UTR_variant	rs1020247920				het	149	43	21610–3–0	10719–2–0	2503–1–0	175–0–0	5847–0–0	593–0–0	987–0–0	0–0–0	786–0–0