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Chr	LeftFlank	RightFlank	ref_allele	mutltype	var_allele	type	Gene_name	Qual	loc_type	consequence	transcript	ref_aa	var_aa	aa_pos	CDPred_scores	dbID	Polyphen_ts	Polyphen_aa	Polyphen_an	CADD_Score	EXAC_POPM	ClinPred
chr9	107555092	107555094	T	SNP	G	nonsynonym	ABCA1		98 exonic	ABCA1:uc004bcl.3:exon42:c.5731A>C:p.I1911I,	uc004bcl.3	I	L	1911	-5 -		uc004bcl.3	I 1911 L	benign	19.77	0	0.78997761
chr1	76205703	76205705	G	SNP	T	nonsynonym	ACADM		100 exonic	ACADM:uc001dgw.4:exon7:c.508G>T:p.A170S,ACADM:uc001dgx.3	uc001dgx.3	A	S	84	-6 -		uc009wbp.2	A 174 S	benign	21.1	0	0.1175106
chr1	76228446	76228454	AAAAAAAA	INDEL	AAAAAAAA	frameshift	a ACADM		25 exonic	ACADM:uc001dgw.4:exon12:c.1265_1271AAAAAAA,AC -		-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr12	121176353	121176355	C	SNP	T	nonsynonym	ACADS		88 exonic	ACADS:uc010ul.1:exon7:c.802C>T:p.R268C,ACADS:uc010ul.3	uc010ul.3	R	C	272	-14 -		uc010ul.1	R 268 C	probably dan	20.8	1.00E-04	0.99944752
chr17	7123240	7123256	GGGCGTGCA	INDEL	GGGCGTGCA	nonframeshi	ACADVL		615 exonic	ACADVL:uc010xtq.2:exon1:c.76_90GGGCGCTGCAGCACG	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr17	7127323	7127325	T	SNP	C	nonsynonym	ACADVL		235 exonic	ACADVL:uc010zxx.4:exon14:c.1337G>C:p.W537C,ACADVL:uc010xtq.1	uc010xtq.1	I	T	467	-8 -		uc010xtq.1	I 480 T	probably dan	8.703	0	0.98384964
chr14	105196229	105196231	A	SNP	C	nonsynonym	ADSSL1		43 exonic	ADSSL1:uc010yge.2:exon1:c.1A>C:p.M1L,	uc010yge.2	M	L	1	2 rs80097179	uc010yge.2	M 1 L	benign	2.92	0	0.00888963
chr1	100336030	100336032	C	SNP	T	nonsynonym	AGL		189 exonic	AGL:uc001dbj.1:exon6:c.740C>T:p.A247V,AGL:uc001dbj.1:exon6:c.740C>T:p.A247V,AGL:uc001dbm.1	uc001dbm.1	A	V	230	0 -		uc001dbm.1	A 230 V	probably dan	33	0	0.9968124
chr1	100347124	100347126	T	SNP	C	nonsynonym	AGL		101 exonic	AGL:uc001dbj.1:exon17:c.2186T>C:p.I729T,AGL:uc001dbm.1:exon17:c.2186T>C:p.I729T,AGL:uc001dbn.1	uc001dbj.1	I	T	729	-1 -		uc001dbm.1	I 713 T	possibly dam	20.7	0	0.91919285
chr1	100353663	100353665	G	SNP	C	nonsynonym	AGL		86 exonic	ccpAGL:uc001dbj.1:exon21:c.2812G>T:p.G938C,AGL:uc001dbm.1:exon21:c.2812G>T:p.G938C,AGL:uc001dbn.1	uc001dbn.1	G	C	921	-3 -		uc001dbn.1	G 921 C	probably dan	24.4	0	0.99944466
chr9	139581758	139581773	CAGCAGCAG	INDEL	CAGCAGCAG	nonframeshi	AGPAT2		312 exonic	AGPAT2:uc004cqj.1:exon1:c.38_51TGCTGCTGCTGCTGCTI-	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr1	976124	976132	AAGAAGA	INDEL	AAGA	nonframeshi	AGRN		17 exonic	AGRN:uc001ack.2:exon4:c.592_598AAGA,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr21	45710527	45710529	G	SNP	C	nonsynonym	AIRE		174 exonic	AIRE:uc001ivx.1:exon1:c.52G>C:p.D18H,	uc001ivx.1	D	H	18	-1 rs2073610	G NaN	NaN	NaN	NaN	3.087	0	None
chr21	45710555	45710560	CCCC	INDEL	CCCCC	frameshift	a AIRE		219 exonic	AIRE:uc001ivx.1:exon1:c.80_83CCCCC,	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr21	45710616	45710618	G	SNP	C	nonsynonym	AIRE		374 exonic	AIRE:uc001ivx.1:exon1:c.141G>C:p.W47C,	uc001ivx.1	W	C	47	-2 -		NaN	NaN	NaN	4.515	0	None
chr21	45710794	45710807	GATGGATGC	INDEL	GA	frameshift	a AIRE		58 exonic	AIRE:uc001ivx.1:exon1:c.319_330GA,AIRE:uc002ez.3:exe -	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr21	45717583	45717585	C	SNP	T	nonsynonym	AIRE		129 exonic	AIRE:uc002ez.3:exon14:c.1612C>T:p.R538C,AIRE:uc033ssu.1:exon1	uc002ez.2	R	C	341	-3 -		uc002ez.2	R 341 C	probably dan	19.65	0	0.99072027
chr9	101980916	101980918	A	SNP	G	nonsynonym	ALG2		150 exonic	ALG2:uc004eaj.3:exon1:c.2717T>C:p.F91L,ALG2:uc004eaf.2:exon1:c.2717T>C:p.F91L,ALG2:uc004eaf.2	uc004eaf.2	F	L	184	-1 -		uc004eaj.2	F 91 L	probably dan	26.7	0	0.98062277
chr9	101984145	101984147	A	SNP	G	nonsynonym	ALG2		141 exonic	ALG2:uc004eaf.3:exon1:c.317C>G:p.S11R,	uc004eaf.2	S	P	11	-1 rs115451437	uc004eaf.2	S 11 P	benign	8.427	0	0.00882664
chr2	202609042	202609044	C	SNP	G	nonsynonym	ALS2		206 exonic	ALS2:uc002uqp.4:exon5:c.2108G>C:p.R703T,ALS2:uc002uqp.4:exon5:c.2108G>C:p.R703T,ALS2:uc002uqp.3	uc002uqp.3	R	T	703	-6 -		uc002uqp.3	R 703 T	probably dan	28.9	0	0.90593332
chr2	202622162	202622164	C	SNP	G	nonsynonym	ALS2		83 exonic	ALS2:uc002uqp.4:exon5:c.1433G>C:p.G478A,ALS2:uc002uqp.2:exon5:c.1433G>C:p.G478A,ALS2:uc002uqp.2	uc002uqp.2	G	A	478	0 -		uc002uqp.2	G 478 A	benign	8.487	0	0.12436493
chr5	34004692	34004694	T	SNP	C	nonsynonym	AMACR		80 exonic	AMACR:uc003jil.2:exon3:c.538A>G:p.I180V,AMACR:uc003jil.1	uc003jil.2	I	V	180	3 -		uc003jil.2	I 180 V	benign	11.23	0	0.18205971
chr11	22249067	22249069	G	SNP	A	nonsynonym	ANO5		117 exonic	ANO5:uc001mqj.2:exon7:c.584G>A:p.R195Q,ANO5:uc001mqj.2:exon7:c.584G>A:p.R195Q,ANO5:uc001mqj.2	uc001mqj.2	R	Q	195	1 -		uc001mqj.2	R 195 Q	possibly dam	26.8	1.00E-04	0.91578249
chr11	22249075	22249077	C	SNP	G	nonsynonym	ANO5		115 exonic	ANO5:uc001mqj.2:exon7:c.592C>G:p.L198V,ANO5:uc001mqj.2:exon7:c.592C>G:p.L198V,ANO5:uc001mqj.2	uc001mqj.2	L	V	197	1 -		uc001mqj.2	L 198 V	possibly dam	18.74	0	0.83588487
chr X	66766356	66766400	GGCGGCGGC	INDEL	GGCGGCGGC	nonframeshi	AR		599 exonic	AR:uc022hp4.1:exon1:c.1369_1420GGCGGCGGCGGCGGCG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr8	1851668	1851670	T	INDEL	TT	frameshift	a ARHGEF10		172 exonic	ARHGEF10:uc003wpt.3:exon13:c.1501_1502delinsTT,	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr8	1871140	1871142	A	SNP	T	nonsynonym	ARHGEF10		63 exonic	ARHGEF10:uc003wps.3:exon18:c.2041A>T:p.M681L,ARI uc003wps.2	uc003wps.2	M	L	452	2 -		uc003wps.2	M 719 L	benign	2.175	0	0.04062302
chr8	1872028	1872030	A	SNP	G	nonsynonym	ARHGEF10		189 exonic	ARHGEF10:uc003wps.3:exon20:c.2363A>G:p.K788R,ARI uc003wps.2	uc003wps.2	R	R	826	2 -		uc003wps.2	R 851 R	probably dan	17.48	0	0.89922547
chr8	17942263	17942265	T	SNP	G	nonsynonym	ASAH1		117 exonic	ASAH1:uc003wyn.2:exon1:c.47G>C:p.R16P,ASAH1:uc003wyn.2:exon1:c.47G>C:p.R16P,ASAH1:uc003wyn.2	uc003wyn.2	R	P	16	-2 -		uc003wyn.2	R 16 P	benign	6.784	0	0.12030354
chr11	63438835	63438837	C	SNP	T	nonsynonym	ATL3		138 exonic	ATL3:uc001nel.1:exon2:c.128G>A:p.C43Y,	uc001nel.1	C	Y	43	-2	rs295988	C uc001nel.1	C 43 Y	benign	0.007	0	0.01208867
chr11	108119734	108119736	A	SNP	G	nonsynonym	ATM		81 exonic	ATM:uc009ver.1:exon10:c.1141A>G:p.S381G,ATM:uc001qbh.1:exon10:c.1141A>G:p.S381G,ATM:uc001qbh.1	uc001qbh.1	G	G	381	0 -		uc001qbh.1	G 381 G	benign	0.085	0	0.04148014
chr11	108175487	108175489	G	SNP	A	nonsynonym	ATM		56 exonic	ATM:uc009ver.1:exon2:c.136G>A:p.A57T,	uc001qbh.1	L	L	513	0 -		NaN	NaN	NaN	6.136	1.00E-04	None
chr6	36306671	36306673	C	SNP	A	nonsynonym	ATXN1		105 exonic	ATXN1:uc003nbt.3:exon8:c.2336G>T:p.S779I,ATXN1:uc003nbt.2:exon8:c.2336G>T:p.S779I,ATXN1:uc003nbt.2	uc003nbt.2	S	I	779	-2 -		uc003nbt.2	S 779 I	benign	14.35	0	0.51067108
chr6	16327201	16327203	C	SNP	T	nonsynonym	ATXN1		140 exonic	ATXN1:uc003nbt.3:exon6:c.1340G>A:p.G447E,ATXN1:uc003nbt.2:exon6:c.1340G>A:p.G447E,ATXN1:uc003nbt.2	uc003nbt.2	G	E	447	-2 -		uc003nbt.2	G 447 E	probably dan	22.5	0	0.8253293
chr6	16327864	16327909	TGCTGCTGCT	INDEL	TGCTGCTGCT	nonframeshi	ATXN1		173 exonic	ATXN1:uc003nbt.3:exon6:c.634_637CAGCAGCAGCAGCA -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	16327896	16327898	C	SNP	A	nonsynonym	ATXN1		140 exonic	ATXN1:uc003nbt.3:exon6:c.645G>T:p.Q215H,ATXN1:uc003nbt.2:exon6:c.645G>T:p.Q215H,ATXN1:uc003nbt.2	uc003nbt.2	Q	H	215	0 -		uc003nbt.2	Q 215 H	unknown	1.752	0	0.00847288
chr6	16327902	16327904	C	SNP	A	nonsynonym	ATXN1		140 exonic	ATXN1:uc003nbt.3:exon6:c.639G>T:p.Q213H,ATXN1:uc003nbt.2:exon6:c.639G>T:p.Q213H,ATXN1:uc003nbt.2	uc003nbt.2	Q	H	213	0 -		uc003nbt.2	Q 213 H	unknown	4.787	0	0.00719906
chr6	16327903	16327921	TGCTGATGC	INDEL	TGCTGATGC	nonframeshi	ATXN1		470 exonic	ATXN1:uc003nbt.3:exon6:c.622_638CAGCAGCATCAGCA -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	16327903	16327909	TGCTG	INDEL	TGCTGCTGA	nonframeshi	ATXN1		49 exonic	ATXN1:uc003nbt.3:exon6:c.634_638CAGCATCAGCAGCA,-	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	16327908	16327910	A	SNP	C	nonsynonym	ATXN1		353 exonic	ATXN1:uc003nbt.3:exon6:c.631T>G:p.H211Q,ATXN1:uc003nbt.2:exon6:c.631T>G:p.H211Q,ATXN1:uc003nbt.2	uc003nbt.2	H	Q	211	0 -		uc003nbt.2	H 211 Q	unknown	5.976	0	0.10039836
chr6	16327909	16327924	TGCTGATGC	INDEL	TGCTGATGC	nonframeshi	ATXN1		388 exonic	ATXN1:uc003nbt.3:exon6:c.619_632CAGCATCAGCA,ATXI -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	16327912	16327918	TGATG	INDEL	TGATGCTG	nonframeshi	ATXN1		491 exonic	ATXN1:uc003nbt.3:exon6:c.625_628CAGCATCA,ATXN1:u -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	16327914	16327916	A	SNP	C	nonsynonym	ATXN1		190 exonic	ATXN1:uc003nbt.3:exon6:c.627T>G:p.H209Q,ATXN1:uc003nbt.2:exon6:c.627T>G:p.H209Q,ATXN1:uc003nbt.2	uc003nbt.2	H	Q	209	0	rs119969612	uc003nbt.2	H 209 Q	benign	0.432	0	0.18909662
chr6	16327915	16327956	TGCTGCTGCT	INDEL	TGCTGCTGCT	nonframeshi	ATXN1		358 exonic	ATXN1:uc003nbt.3:exon6:c.587_626AGCAGCAGCAGCAG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	16327915	16327921	TGCTG	INDEL	TG	nonframeshi	ATXN1		474 exonic	ATXN1:uc003nbt.3:exon6:c.622_628CA,ATXN1:uc003jsh.-	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr22	46067965	46067967	C	SNP	T	nonsynonym	ATXN10		28 exonic	ATXN10:uc003bgm.2:exon1:c.23C>T:p.P8L,ATXN10:uc003bgm.1	uc003bgm.1	P	L	8	-3 -		uc003bgm.1	P 8 L	possibly dam	12.67	1.00E-04	None
chr22	46125390	46125400	T	INDEL	TTTTTTTTTT	frameshift	a ATXN10		135 exonic	ATXN10:uc003bgm.2:exon7:c.824T>C:p.I275T,ATXN10:uc003bgm.1	uc003bgm.1	T	I	275	-1 -		uc003bgm.1	I 275 T	benign	8.304	1.00E-04	None
chr12	112036753	112036782	GCTGCTGCTI	INDEL	GCTGCTGCTI	nonframeshi	ATXN2		191 exonic	ATXN2:uc001tuj.3:exon1:c.538_565CAGCAGCAGCAGCAG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr12	112036767	112036797	TGCTGCTGCT	INDEL	TGCTGCTGCT	nonframeshi	ATXN2		771 exonic	ATXN2:uc001tuj.3:exon1:c.523_551CAACAGCAGCAGCA, -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr12	112036965	112036967	G	SNP	A	nonsynonym	ATXN2		118 exonic	ATXN2:uc001tuj.3:exon1:c.353C>T:p.P118L,	uc001tuj.2	P	L	118	-3 -		uc001tuj.2	P 118 L	benign	11.25	0	0.25400221
chr14	92537353	92537355	C	SNP	G	nonsynonym	ATXN3		201 exonic	ATXN3:uc021ron.1:exon10:c.916G>C:p.E306R,ATXN3:uc010avh.2	G	R	240	-2	rs128953570	uc001yac.3	G 306 R	benign	9.58	0	0.6072396
chr14	92537353	92537354	T	INDEL	GCTGCTGCTG	nonframeshi	ATXN3		139 exonic	ATXN3:uc021ron.1:exon10:c.916_917insGCAGCAGCAGCAG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr14	92537354	92537379	CTGCTGCTGI	INDEL	CTGCTGCTGI	nonframeshi	ATXN3		307 exonic	ATXN3:uc021ron.1:exon10:c.892_915CAGCAGCAGCAGCAG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr14	92537364	92537387	TGCTGCTGCT	INDEL	TGCTGCTGCT	nonframeshi	ATXN3		391 exonic	ATXN3:uc021ron.1:exon10:c.884_905AGCAGCAACAGCAG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr3	63898319	63898336	GCGCGCGCG	INDEL	GCGCGCGCG	nonframeshi	ATXN7		22 exonic	ATXN7:uc003ebv.4:exon3:c.46_61GCGCGCGCGCGCGCG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr3	63898360	63898393	GCAGCAGCA	INDEL	GCAGCAGCA	nonframeshi	ATXN7		750 exonic	ATXN7:uc003ebv.4:exon3:c.87_118GCAGCAGCAGCAGCAGC -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr3	63898389	63898391	A	SNP	C	nonsynonym	ATXN7		35 exonic	ATXN7:uc003ebv.4:exon3:c.116A>C:p.Q39P,ATXN7:uc003ebv.3	uc003ebv.3	Q	P	39	-1 -		uc003ebv.2	Q 39 P	unknown	6.467	0	0.03082094
chr3	63898390	63898393	GC	INDEL	GCAGCAGCC	nonframeshi	ATXN7		13 exonic	ATXN7:uc003ebv.4:exon3:c.117_118GCAGCAGCCGCCATG -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr3	63967899	63967901	A	SNP	G	nonsynonym	ATXN7		293 exonic	ATXN7:uc003ebv.4:exon7:c.781A>G:p.K261R,ATXN7:uc003ebv.2	uc003ebv.2	K	R	264	2	rs105333188	A uc003ebv.1	K 219 R	probably dan	16.9	0	0.02421619
chr12	58023948	58023950	T	SNP	C	nonsynonym	B4GALNT1		219 exonic	B4GALNT1:uc001qqg.2:exon6:c.698A>G:p.N233S,B4G AL uc010ovr.1	uc010ovr.1	N	S	233	1 -		uc001sph.2	N 233 S	possibly dam	11.25	0	0.35517764
chr9	95480096	95480098	C	SNP	T	nonsynonym	BICD2		75 exonic	BICD2:uc004ssp.1:exon6:c.224G>A:p.R747H,BICD2:uc004ass.1	uc004ass.1	R	H	747	0 -		uc004ass.1	R 747 H	probably dan	32	0	0.99854004
chr2	127827605	127827607	T	SNP	C	nonsynonym	BIN1		140 exonic	BIN1:uc010ysg.2:exon5:c.376A>G:p.M126V,BIN1:uc003ezb.1	uc010ysg.1	M	V	126	-5 -		uc003ezb.1	M 126 V	benign	15.23	0	0.39668006
chr10	102747992	102747994	A	SNP	C	nonsynonym	C10orf92		140 exonic	C10orf2:uc001kxf.2:exon1:c.26A>C:p.Y9S,C10orf2:uc001kxf.2	uc001kxf.2	Y	S	9	-2 -		uc001kxf.2	Y 9 S	benign	3.456	0	0.1835385
chr10	102748011	102748013	A	SNP	C	nonsynonym	C10orf92		140 exonic	C10orf2:uc001kxf.2:exon1:c.45A>C:p.L15F,C10orf2:uc001kxf.2	uc001kxf.2	L	F	15	0 -		uc001kxf.2	L 15 F	probably dan	12.75	0	0.58502692
chr1	200310098	200310100	G	SNP	T	nonsynonym	CACNA1S		140 exonic	CACNA1S:uc001gxn.3:exon24:c.3026C>A:p.T1009K,	uc001gxn.2	T	K	1009	-2 -		uc001gxn.2	T 1009 K	probably dan	33	0	0.99662265
chr1	201035375	201035377	T	SNP	C	nonsynonym	CACNA1S		122 exonic	CACNA1S:uc001gxn.3:exon21:c.2726A>G:p.N909S,	uc001gxn.2	N	S	909	2 -		uc001gxn.2	N 909 S	possibly dam	18.9	0	0.53944581
chr1	201052334	201052336	C	SNP	T	nonsynonym	CACNA1S		136 exonic	CACNA1S:uc001gxn.3:exon10:c.1348G>A:p.A450T,	uc001gxn.2	A	T	450	0 -		uc001gxn.2	A 450 T	probably dam	29.6	1.00E-04	0.57260972
chr15	42697152	42697154	C	SNP	T	splicing	CAPN3		115 splicing	-	-	-	-	-	-30 -		NaN	NaN	NaN	4.328	0	None
chr20	44801491	44801493	A	SNP	C	nonsynonym	CDH22		101 exonic	CDH22:uc010qfd.2:exon12:c.2140T>G:p.S714A,	uc010qfd.1	S	A	714	2 -		uc002wm.2	S 714 A	benign	5.943	0	0.01787779
chr11	2906196	2906236	CTGCGCACCG	INDEL	CCGCGCACCG	nonframeshi	CDKN1C		166 exonic	CDKN1C:uc009vdf.3:exon2:c.452_490CGCGCTCCGGTCGCC -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr10	50822259	50822261	A	SNP	G	nonsynonym	CHAT		31 exonic	CHAT:uc001ytv.2:exon1:c.25A>G:p.R9G,	uc001ytv.2	R	G	9	-2 -		uc001ytv.2	R 9 G	benign	11.23	0	0.02532788
chr10	50822391	50822393	G	SNP	A	nonsynonym	CHAT		100 exonic	CHAT:uc001ytv.2:exon1:c.157G>A:p.G53R,	uc001ytv.2	G	R	53	-2 -		uc001ytv.2	G 53 R	benign	8.771	0	0.03655029
chr8	61693942	61693952	AAAGCAAAA	INDEL	AAAGCAAAA	nonframeshi	CHD7		188 exonic	CHD7:uc003eue.3:exon3:c.2050_2058AAAGCAAAAGCAA -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr8	61741305	61741307	C	SNP	T	nonsynonym	CHD7		123 exonic	CHD7:uc003eue.3:exon14:c.3463C>T:p.R1155C,	uc003eue.2	R	C	1155	-7 -		uc003eue.2	R 1155 C	probably dam	28.9	1.00E-04	0.89888185
chr8	61761698	61761700	G	SNP	C	nonsynonym	CHD7		91 exonic	CHD7:uc003eue.3:exon25:c.5390G>C:p.G1797A,	uc003eue.2	G	A	1797	0 -		uc003eue.2	G 1797 A	probably dam	19.04	0	0.96501952
chr2	175676256	175676258	T	SNP	C	nonsynonym	CHN1		77 exonic	CHN1:uc002ug.3:exon9:c.271A>G:p.K91E,CHN1:uc010bxe.uc002ug.2	uc002ug.2	K	E	91	-6 -		uc010bxq.1	K 290 E	probably dan	33	0	0.95453489
chr2	233930247	233930249	G	SNP	T	nonsynonym	CHRND		140 exonic	CHRND:uc002xxw.4:exon5:c.391G>T:p.V131L,CHRND:uc010bmg.1	uc010bmg.1	V	L	116	-1 -		uc002xxw.2	V 131 L	probably dam	19.03	0	0.88519377
chr2	233935681	233935683	G	SNP	A	splicing	CHRND		59 splicing	uc003rzn.1:exon6:c.316+1G>A:uc002xxw.4:exon6:c.619+ -	-	-	-	-	-30 -		NaN	NaN	NaN	14.94	0	None
chr7	4806321	4806323	C	SNP	T	nonsynonym	CHRNE		336 exonic	CHRNE:uc002tk1.1:exon1:c.370>A:p.G118T,	uc002tk1.1	G	R	13	-2 -		uc002tk1.1	G 13 R	benign	11.28	1.00E-04	0.13646635
chr3	9908961	9908963	G	SNP	A	nonsynonym	CIDEC		98 exonic	CIDEC:uc010fcp.3:exon3:c.226C>T:p.P76S,	uc010fcp.2	P	S	76	-1 -		uc010fcp.2	P 76 S	benign	8.355	0	0.07031593
chr7	14801421	14801423	T	SNP	G	nonsynonym	CLCN1		38 exonic	CLCN1:uc003wcr.1:exon11:c.1117T>G:p.N39K,	uc003wcr.1	N	K	39	0 -		uc003wcr.1	N 39 K	benign	12.55	0	0.09911196
chr16	28493828	28493830	T	SNP	C	nonsynonym	CLN3		273 exonic	CLN3:uc002dps.1:exon7:c.494A>G:p.Y165C,CLN3:uc010ir uc002dps.1	uc002dps.1	Y	C	139	-11 -		uc002dps.1	Y 238 C	probably dan	17.13	1.00E-04	0.99956197
chr16	28500680	28500682	G	SNP	A	nonsynonym	CLN3		107 exonic	CLN3:uc010bpe.1:exon3:c.152C>T:p.S51F,CLN3:uc002dps uc010bpe.1	uc002dps.1	S	F	51	-3 -		uc002dps.1	S 51 F	probably dan	24.1	0	0.94148125
chr16	28502843	28502845	C	SNP	A	stopgain_SN	CLN3		140 exonic	CLN3:uc002dps.1:exon2:c.890>A:p.W30X,	uc002dps.1	W	*	30	-30 -		NaN	NaN	NaN	9.15	0	None
chr11	57426925	57426928	GG	INDEL	G	splicing	CLP1		179 splicing	-	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr3	1363439	1363441	G	SNP	T	stopgain_SN	CNTN6		92 exonic	CNTN6:uc003bpa.3:exon8:c.868G>T:p.E290X,CNTN6:uc003bpa.2	uc003bpa.2	E	*	290	-30 -		NaN	NaN	NaN	44	0	None
chr6	75855167	75855169	G	SNP	C	nonsynonym	COL12A1		80 exonic	COL12A1:uc021bse.1:exon9:c.1072C>T:p.R358C,COL12A uc003phs.2	uc003phs.2	R	C	358	-6 -		uc003phs.2	R 1522 C	probably dam	14.49	1.00E-04	0.94988334
chr6	75861863	75861865	C	SNP	G	nonsynonym	COL12A1		144 exonic	COL12A1:uc021bse.1:exon3:c.326G>C:p.G109A,COL12A uc003phs.2	uc003phs.2	G	A	109	-7 -		uc003phs.2	G 109 A	probably dan	22.5	1.00E-04	0.94561964
chr10	71662527	71662529	C	SNP	T	nonsynonym	COL13A1		129 exonic	COL13A1:uc010ysz.1:exon14:c.739C>G:p.Q247E,COL13A uc010ysz.1	uc010ysz.1	Q	E	269	2 -		uc010ysz.1	Q 247 E	probably dan	18.95	0	0.7468169
chr21	47410908	47410910	C	SNP	T	nonsynonym	COL6A1		105 exonic	COL6A1:uc002ruu.1:exon15:c.1073C>T:p.P358L,	uc002ruu.1	P	L	358	-11 -		uc002ruu.1	P 358 L	probably dam	16.66	0	0.98278481
chr21	47412310	47412312	G	SNP	A	nonsynonym	COL6A1		140 exonic	COL6A1:uc002ruu.1:exon18:c.1271G>A:p.R424Q,	uc002ruu.1	R	Q	424	1 -		uc002ruu.1	R 424 Q	probably dam	17.4	1.00E-04	0.41706696
chr21	47532185	47532187	G	SNP	A	nonsynonym	COL6A2		140 exonic	COL6A2:uc002rhz.1:exon3:c.409G>A:p.A137T,COL6A2:uc002rhz.1	uc002rhz.1	A	T	137	-8 -		uc002rhz.1	A 137 T	probably dan	18.55	0	0.9658531
chr21	47551953	47551955	C	SNP	T	nonsynonym	COL6A2		174 exonic	COL6A2:uc002rza.1:exon28:c.2548C>T:p.H850Y,	uc002rza.1	H	Y	850	-3 -		uc002rza.1	H 850 Y	benign	10.78	1.00E-04	0.14369014
chr2	238244878	238244878	GCAGCAGCA	INDEL	GCAGCAGCA	nonframeshi	COL6A3		345 exonic	COL6A3:uc002vwx.2:exon39:c.8248_8261GCTGCTGCTGCTGI -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr2	238261208	238261210	C	SNP	G	nonsynonym	COL6A3		62 exonic	COL6A3:uc002vwp.1:exon3:c.172G>C:p.G58R,COL6A3:uc002vwp.1	G	R	58	-2		uc010ynj.1	G 1630 R	probably dan	13.74	0	0.9857502
chr2	238263603	238263605	G	INDEL	"	nonframeshi	d COL6A3		0 exonic	COL6A3:uc002vwp.1:exon3:c.28delCT:p.D10fs,	NaN	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr2	238280405	238280411	CTTCT	INDEL	CT	nonframeshi	d COL6A3		71 exonic	COL6A3:uc002vwo.2:exon8:c.3632_3636AG,COL6A3:uc0 -	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr20	61455814	61455829	CCAGGTCCCT	INDEL	CCAGG	nonframeshi	COL9A3		316 exonic	COL9A3:uc002yelm.3:exon11:c.538_551CCAGG,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr3	15520887	15520889	C	SNP	T	nonsynonym	COL7A1		151 exonic	splicingCOL7A1:uc003bbb.3:exon44:c.3230>A:p.G1080.COL7A1:uc010 uc010hee.2	G	G	1080	-3		uc003bzz.2	G 108 D	probably dan	10.2	0	0.98729521
chr17	13977740	13977742	A	SNP	G	nonsynonym	COX10		93 exonic	COX10:uc002gcf.4:exon2:c.145A>G:p.T49A,	uc002gcf.3	T	A	49	0 -		uc002gcf.3	T 49 A	possibly dam	17.72	0	0.96179175
chr10	101488614	101488616	G	SNP	C	nonsynonym	COX15		140 exonic	COX15:uc003ezc.4:exon4:c.492C>G:p.I164M,COX15:uc003ezb.1	uc003ezb.1	M	I	164	-5 -		uc003ezb.1	I 164 M	benign	14.41	0	0.18122095
chr12	120876261	120876263	C	SNP	T	nonsynonym	COX6A1		140 exonic	COX6A1:uc001ytf.2:exon2:c.184C>T:p.H62Y,	uc001ytf.1	H	Y	62	-7 -		uc001ytf.1	H 62 Y	benign	15.36	0	0.19621912
chr1	53678968	53678970	G	SNP	A	nonsynonym	CP72		82 exonic	CP72:uc001ovb.4:exon5:c.167G>A:p.R56Q,	uc001ovb.3	R	Q	560	-5 -		uc001ovb.3	R 560 Q	probably dam	19.31	1.00E-04	0.120353
chr2	36726453	36726455	G	SNP	A	nons

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Chr	LeftFlank	RightFlank	ref_allele	mutype	var_allele	type	Gene_name	Qual	loc_type	consequence	transcript	ref_aa	var_aa	aa_pos	CDPred_scores	dbID	Polyphen_ts	Polyphen_aa	Polyphen_an	CADD_Score	EXAC_POPMA	ClinPred
chr1	154965149	154965153	GTG	INDEL	G	splicing	FLAD1		52 splicing	-	-	-	-	-	-	-	NaN	NaN	NaN	NaN	0	0.83973229
chr7	128484191	128484193	G	SNP	A	nonsynonym	FLNC		112 exonic	FLNC:uc003xxa.4:exon20:c.30646G>A:p.E10224K,FLNC:uc0	uc003xxa.3	E	K	1022	3 -		uc003xxa.3	E 1022 K	benign	16.76	0	0.83973229
chr1	63884588	63884590	C	SNP	T	nonsynonym	FLRT1		140 exonic	FLRT1:uc021qbx.1:exon1:c.850C>T:p.H284Y,FLRT1:uc001	uc001rxy.1	H	Y	284	-8 -		uc001rxy.1	H 256 Y	benign	7.686	0	0.71538163
chr1	63884901	63884903	C	SNP	T	nonsynonym	FLRT1		140 exonic	FLRT1:uc021qbx.1:exon1:c.1163C>T:p.A388V,FLRT1:uc001	uc001rxy.1	A	V	388	0 -		uc001rxy.1	A 360 V	benign	0.006	0	0.03284728
chrX	147011477	147011479	A	SNP	G	nonsynonym	FMR1		307 exonic	FMR1:uc022qc.2:exon6:c.431A>G:p.E144G,FMR1:uc00	uc004fc1.3	E	G	5	-2 -		uc011mwz.1	E 144 G	possibly damaging	17.38	0	None
chr17	78086700	78086702	G	SNP	A	nonsynonym	GAA		114 exonic	GAA:uc002zxq.3:exon14:c.1915G>A:p.V639M,GAA:uc00	uc002zxq.2	V	M	639	4 -		uc002zxq.2	V 639 M	possibly damaging	19.67	0	0.05218599
chr7	30671913	30671915	G	SNP	C	nonsynonym	GAR5		99 exonic	GAR5:uc003thm.2:exon16:c.1955G>C:p.G652A	uc003thm.2	G	A	652	-7 -		uc003thm.2	G 652 A	benign	21.3	0	0.06315676
chr9	35740888	35740890	C	SNP	T	nonsynonym	GBA2		140 exonic	GBA2:uc011qxd.2:exon6:c.977G>A:p.R326Q,GBA2:uc001	uc003xxy.1	R	Q	33	1 -		uc003xxy.1	R 326 Q	benign	1.053	1.00E-04	0.0219721
chr3	81699025	81699027	G	SNP	A	nonsynonym	GBE1		117 exonic	GBE1:uc021aax.1:exon4:c.353C>T:p.P118L,GBE1:uc021a	uc003dgg.2	P	L	159	-14 -		uc003dgg.2	P 159 L	probably damaging	29.6	0	0.9939785
chr2	69554079	69554081	A	SNP	G	nonsynonym	GFPT1		170 exonic	GFPT1:uc002vfr.2:exon19:c.2021T>C:p.L674P,GFPT1:uc0	uc002vfr.2	L	P	656	-10 -		uc002vfr.2	L 656 P	probably damaging	23.5	0	None
chr9	124044745	124044747	C	SNP	T	nonsynonym	GSN		179 exonic	GSN:uc010mvu.2:exon3:c.17C>T:p.T6M,	uc010mvu.2	T	M	6	-1 -		NaN	NaN	NaN	5.155	0	None
chr9	124045670	124045672	G	SNP	A	splicing	GSN		35 splicing	-	-	-	-	-	-30 -		NaN	NaN	NaN	0.206	0	None
chr9	124073046	124073048	G	SNP	T	nonsynonym	GSN		172 exonic	GSN:uc010mvt.1:exon6:c.487G>T:p.D163Y,GSN:uc004bl	uc010mvt.1	D	Y	163	-10 -		uc011yfu.1	D 171 Y	probably damaging	22.2	0	0.99975592
chr3	148714546	148714548	G	SNP	C	nonsynonym	GYG1		83 exonic	GYG1:uc003ewn.3:exon4:c.337G>C:p.D113H,GYG1:uc00	uc003ewn.2	D	H	113	-4 -		uc003ewn.2	D 113 H	probably damaging	19.01	0	0.94648731
chr2	26477125	26477130	ACTACTA	INDEL	ACTACTA	nonsynonym	HAOX1		690 exonic	HAOX1:uc002ucq.3:exon12:c.4_7ACTACTA,HAOX1:uc002uc	-	-	-	301	-		NaN	NaN	NaN	NaN	0	None
chr6	26093021	26093023	T	SNP	G	nonsynonym	HFE		153 exonic	HFE:uc003ngx.1:exon2:c.186T>G:p.D62E,HFE:uc003ngb.	uc003nfc.1	D	E	154	-5 -		uc003ngc.1	D 150 E	probably damaging	12.72	0	0.87574619
chr6	26093408	26093410	A	SNP	T	nonsynonym	HFE		110 exonic	HFE:uc003ngx.1:exon3:c.415A>T:p.I139F,HFE:uc003ngb.	uc003nfc.1	I	F	319	0 -		uc003ngx.1	I 139 F	possibly damaging	16.86	0	0.28593346
chr10	71060523	71060525	G	SNP	A	splicing	HK1		140 splicing	-	-	-	-	-	-30	rs571118526	NaN	NaN	NaN	7.5	0	None
chr2	176981660	176981662	A	SNP	G	nonsynonym	HOXD10		99 exonic	HOXD10:uc002ukj.3:exon1:c.100A>G:p.M34V,	uc002ukj.2	M	V	34	1 -		uc002ukj.2	M 34 V	benign	5.328	0	0.75605494
chr2	176981958	176981960	GGTCTGAGG	INDEL	GGTCTGAGG	frameshift_p	HOXD10		239 exonic	HOXD10:uc002ukj.3:exon2:c.1023_1048delGTCTGAGGCCC	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr1	22181218	22181220	G	SNP	A	nonsynonym	HSPG2		123 exonic	HSPG2:uc001bfj.3:exon49:c.6173C>T:p.S2058L,HSPG2:uc	uc001bfj.2	S	L	2058	-5 -		uc001bfj.2	S 2058 L	probably damaging	14.77	1.00E-04	0.68937945
chr1	22206682	22206684	C	SNP	T	nonsynonym	HSPG2		140 exonic	HSPG2:uc001bfj.3:exon17:c.2260G>A:p.G754S,HSPG2:uc	uc001bfj.2	G	S	755	0 -		uc001bfj.2	G 754 S	benign	12.44	0	0.10509239
chr7	112102104	112102106	A	SNP	G	nonsynonym	IFRD1		123 exonic	IFRD1:uc003wqk.3:exon3:c.668A>G:p.Y223C,IFRD1:uc00	uc003wqj.2	Y	C	223	-9 -		uc003wqj.2	Y 223 C	benign	13.59	0	0.91550446
chr7	112107982	112107985	GGT	INDEL	''	frameshift_d	IFRD1		0 exonic	IFRD1:uc003wqk.3:exon1:c.5_6del:p.2_2del,	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr7	112107984	112107986	T	SNP	A	nonsynonym	IFRD1		85 exonic	IFRD1:uc003wqk.3:exon1:c.7T>A:p.S3T,	uc003wqk.2	S	T	3	1	rs116076119	NaN	NaN	NaN	6.361	0	None
chr7	112107985	112107987	C	SNP	A	stopgain_SN	IFRD1		80 exonic	IFRD1:uc003wqk.3:exon1:c.8C>A:p.S3X,	uc003wqk.2	S	*	3	-30	rs114489785	NaN	NaN	NaN	3.513	0	None
chr11	68697795	68697797	A	SNP	G	nonsynonym	IGHMBP2		65 exonic	IGHMBP2:uc001oof.1:exon1:c.1A>G:p.M1V,	uc001oof.1	M	V	1	1 -		NaN	NaN	NaN	1.17	1.00E-04	None
chr14	105156075	105156077	G	SNP	A	splicing	INF2		231 splicing	-	-	-	-	-	-30	rs115602636	NaN	NaN	NaN	13.59	0	None
chr14	105173862	105173890	CCCCACCCCC	INDEL	CCCCACCCCCC	nonframeshi	INF2		220 exonic	INF2:uc001yeb.2:exon8:c.1259_1285CCCCACCCCCACCCC,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr14	105174284	105174286	A	SNP	C	nonsynonym	INF2		26 exonic	INF2:uc001yeb.2:exon8:c.1681A>C:p.T561P,INF2:uc001y	uc001yeb.2	T	P	561	1 -		uc001yeb.2	T 561 P	possibly damaging	12.58	0	0.90142298
chr14	105181652	105181654	A	SNP	G	nonsynonym	INF2		99 exonic	INF2:uc001yeb.2:exon22:c.3727A>G:p.K1243E,	uc001yeb.2	K	E	1243	1 -		uc001yeb.2	K 1243 E	benign	5.183	0	0.03153324
chr12	56089054	56089056	T	SNP	A	nonsynonym	ITGA7		140 exonic	ITGA7:uc009bxm.3:exon22:c.1597G>T:p.A533S,ITGA7:uc0	uc003ubq.2	A	S	652	2 -		uc003ubq.2	A 755 S	possibly damaging	13.18	0	0.19797568
chr12	56596536	56596538	G	SNP	A	nonsynonym	ITPR2		154 exonic	ITPR2:uc009bjg.1:exon9:c.842C>T:p.S281L,ITPR2:uc001d	uc009bjg.1	S	L	281	-2 -		uc001rhg.2	S 2130 L	benign	11.39	1.00E-04	0.04705433
chr12	26753040	26753042	T	SNP	C	nonsynonym	ITPR2		65 exonic;splice	ITPR2:uc001rhg.3:exon29:c.3680A>G:p.N12275,	uc001rhg.2	N	S	1227	-1 -		uc001rhg.2	N 1227 S	benign	2.582	0	0.01112172
chr12	26834804	26834807	AC	INDEL	ACTCAC	splicing	ITPR2		421 splicing	-	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr1	10386241	10386243	G	SNP	A	nonsynonym	KIF1B		173 exonic	KIF1B:uc001wfb.3:exon25:c.2707G>A:p.D903N,KIF1B:uc0	uc001aqz.2	D	N	917	1 -		uc001aqx.3	D 917 N	probably damaging	25.8	1.00E-04	0.37034601
chr17	4923885	4923887	C	SNP	T	nonsynonym	KIF1C		132 exonic	KIF1C:uc002qzm.2:exon20:c.1850C>T:p.P617L,	uc002qzm.1	P	L	617	-3 -		uc002qzm.1	P 617 L	benign	18.81	0	0.44532934
chr12	39716494	39716496	C	SNP	A	nonsynonym	KIF21A		94 exonic	KIF21A:uc001fxe.3:exon13:c.1597G>T:p.G533C,KIF21A:u	uc001fxd.1	G	C	221	-3 -		uc001fxd.1	G 1196 C	probably damaging	18.45	0	0.93174136
chr12	39724565	39724567	G	SNP	T	nonsynonym	KIF21A		88 exonic	KIF21A:uc001fxe.3:exon9:c.1252C>A:p.L418I,KIF21A:uc0	uc001fxe.2	L	I	113	2 -		uc001fxe.2	L 1065 I	probably damaging	12.87	1.00E-04	0.27533039
chr12	39726879	39726879	A	SNP	G	nonsynonym	KIF21A		201 exonic	KIF21A:uc001fxe.3:exon5:c.470T>C:p.M157T,KIF21A:uc0	uc001fxe.2	M	T	840	-3 -		uc001fxe.2	M 827 T	probably damaging	11.69	1.00E-04	0.10268153
chr12	57974933	57974935	C	SNP	T	nonsynonym	KIF5A		127 exonic	KIF5A:uc010nr.1:exon21:c.2467C>T:p.R823W,KIF5A:uc0	uc010nr.1	R	W	823	-3 -		uc010nr.1	R 823 W	probably damaging	20.2	0	0.97465366
chr1	170010856	170010858	AA	INDEL	-	nonframeshi	KIFAP3		60 exonic	KIFAP3:uc010ghy.2:exon4:c.45_46T,	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr3	42733380	42733390	GAGGAGGAG	INDEL	GAGGAG	nonframeshi	KLHL40		127 exonic	KLHL40:uc003clv.1:exon6:c.1762_1770GAGGAG,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr6	129371233	129371235	G	SNP	A	splicing	LAMA2		222 splicing	uc003qbh.3:exon2:c.283+1G>A,uc003qbh.3:exon2:c.283-	-	-	-	-	-30 -		NaN	NaN	NaN	21.5	1.00E-04	None
chr6	129498007	129498009	G	SNP	A	nonsynonym	LAMA2		117 exonic	LAMA2:uc021sfb.1:exon30:c.1364G>A:p.R455Q,LAMA2:uc003qbh.2	uc003qbh.2	R	Q	455	1 -		uc003qbh.2	R 455 Q	possibly damaging	24.2	1.00E-04	0.74790865
chr6	129612839	129612841	A	SNP	G	nonsynonym	LAMA2		106 exonic	LAMA2:uc021sfb.1:exon20:c.2831A>G:p.Q944R,LAMA2:u	uc003qbh.2	Q	R	944	-3 -		uc003qbh.2	Q 944 R	benign	12.1	1.00E-04	0.19153166
chr6	129636904	129636906	A	SNP	G	splicing	LAMA2		122 splicing	uc003qbh.3:exon2:c.3736-2A>T,uc003qbo.3:exon26:c.3	-	-	-	-	-30 -		NaN	NaN	NaN	22.3	0	None
chr6	129802438	129802440	G	SNP	A	nonsynonym	LAMA2		109 exonic	LAMA2:uc021sfb.1:exon55:c.7604G>A:p.G2535D,LAMA2:u	uc003qbn.2	G	D	2533	-3 -		uc003qbn.2	G 2535 D	probably damaging	27	1.00E-04	0.86261159
chr5	49160986	49160988	T	SNP	C	nonsynonym	LAMB2		140 exonic	LAMB2:uc003cwe.3:exon25:c.3875A>G:p.N1292S,	uc003cwe.2	N	S	1292	1 -		uc003cwe.2	N 1292 S	benign	9.951	0	0.19659663
chr5	49161383	49161385	C	SNP	A	nonsynonym	LAMB2		193 exonic	LAMB2:uc003cwe.3:exon24:c.3574G>T:p.A1192S,	uc003cwe.2	A	S	1192	1 -		uc003cwe.2	A 1192 S	benign	10.45	0	0.04267314
chrX	64749509	64749511	G	SNP	A	nonsynonym	LAS1L		586 exonic	LAS1L:uc004wxd.2:exon4:c.637C>T:p.H213Y,LAS1L:uc00	uc004wxc.1	H	Y	255	2 -		uc004wxa.1	H 255 Y	benign	3.116	0	None
chr11	18418147	18418150	CT	INDEL	C	frameshift_d	LDHA		81 exonic	LDHA:uc010rdd.2:exon1:c.54_55C,	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr22	30639773	30639775	A	SNP	G	nonsynonym	LIF		348 exonic	LIF:uc003agz.3:exon3:c.475T>C:p.Y159H,	uc003agz.2	Y	H	159	-6 -		uc003agz.2	Y 159 H	probably damaging	21.8	0	None
chr1	156105033	156105035	C	SNP	T	nonsynonym	LMNA		140 exonic	LMNA:uc009wrp.3:exon2:c.50C>T:p.T17M,	uc009wrp.2	M	T	17	-1 -		NaN	NaN	NaN	9.888	1.00E-04	None
chr19	2431797	2431799	C	SNP	T	nonsynonym	LMNB2		112 exonic	LMNB2:uc002ky.4:exon10:c.1693G>C:p.V565L,	uc002kwu.1	V	L	565	0 -		uc002ryv.2	V 545 L	benign	7.416	1.00E-04	0.58186096
chr3	69171496	69171502	AGAAG	INDEL	AG	nonframeshi	LMOD3		126 exonic	LMOD3:uc003dnt.2:exon2:c.37_41CT,LMOD3:uc003dns.1	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr11	46896614	46896618	T	SNP	A	nonsynonym	LRP4		82 exonic	LRP4:uc010ehd.4:exon28:c.3965A>C:p.H1322P,	uc010ehd.3	H	P	1322	-2 -		uc010ehd.3	H 1322 P	probably damaging	23	1.00E-04	0.60312916
chr1	235918940	235918942	C	SNP	G	nonsynonym	LYST		58 exonic	LYST:uc001hkj.3:exon25:c.7066G>C:p.D2356H,	uc001hkj.2	D	H	2356	-1 -		uc001hkj.2	D 2356 H	benign	11.39	0	0.26023567
chr1	235922404	235922406	C	SNP	T	nonsynonym	LYST		140 exonic	LYST:uc001hkj.3:exon23:c.6658G>A:p.D2220N,	uc001hkj.2	D	N	2220	1 -		uc001hkj.2	D 2220 N	benign	16.6	0	0.86049976
chr1	235972993	235972995	G	SNP	A	nonsynonym	LYST		180 exonic	LYST:uc001hkl.2:exon5:c.1124C>T:p.A375V,LYST:uc001k	uc001hkl.1	A	V	375	0 -		uc001hkl.1	A 375 V	benign	12.04	1.00E-04	0.13634152
chr11	47308015	47308017	A	SNP	G	nonsynonym	MAAT		62 exonic	MAAD:uc001neq.2:exon15:c.2584A>G:p.S862G,MAAD:u	uc001ner.1	G	G	862	0 -		uc009yln.1	S 819 G	probably damaging	28.6	0	0.56126522
chr17	44039818	44039820	C	SNP	T	nonsynonym	MAPT		195 exonic	MAPT:uc002zxq.3:exon2:c.116C>T:p.T39M,MAPT:uc002zq.	uc002zq.3	T	M	39	-1 -		uc002zq.3	T 39 M	benign	4.542	1.00E-04	0.01297364
chr17	44061122	44061124	C	SNP	T	nonsynonym	MAPT		276 exonic	MAPT:uc002zxq.3:exon11:c.497C>T:p.S166L,MAPT:uc010d	uc010dau.2	S	L	318	-2	rs73314997	uc010dau.2	S 318 L	benign	14.68	0	0.01975875
chr17	44071865	44071867	C	SNP	T	nonsynonym	MAPT		66 exonic	MAPT:uc002zxq.3:exon7:c.571A>C:p.T191P,MAPT:uc002	uc002zq.3	T	P	537	-1 -		uc002zq.3	T 191 P	probably damaging	8.256	0	0.58954542
chr5	138641509	138641511	G	SNP	A	nonsynonym	MATR3		203 exonic	MATR3:uc010jfb.3:exon4:c.406G>A:p.E136K,MATR3:uc0	uc003ldu.2	E	K	136	1 -		uc003ldu.2	E 136 K	probably damaging	20.4	0	0.90659624
chr1	29522472	29522474	T	SNP	C	stopgain_SN	MECR		181 exonic	MECR:uc001qrp.1:exon9:c.716G>A:p.W239X,MECR:uc00	uc001ber.1	W	*	239	-30 -		NaN	NaN	NaN	37	0	None
chr1	29557360	29557362	G	SNP	A	nonsynonym	MECR		19 exonic	MECR:uc010afz.1:exon1:c.58C>G:p.L20V,MECR:uc010ao	uc010afz.1	L	V	20	1 -		uc010ntq.1	L 20 V	benign	15.89	0	0.04629333
chr1	12052716	12052718	G	SNP	T	nonsynonym	MFN2		122 exonic	MFN2:uc009mi.3:exon3:c.281G>T:p.R94L,MFN2:uc001a	uc009mi.2	L	L	94	-2 -		uc001atn.3	R 94 L	probably damaging	31	0	0.98917108
chr1	12062084	12062086	C	SNP	G	nonsynonym	MFN2		277 exonic	MFN2:uc009mi.3:exon10:c.1385C>G:p.T462R,MFN2:uc00	uc001atn.3	T	R	362	-1 -		uc001atn.3	T 362 R	possibly damaging	20.7	0	0.59035028
chr2	190927060	190927062	G	SNP	A	nonsynonym	MSTN		308 exonic	MSTN:uc002urp.3:exon1:c.262C>T:p.R88W,	uc002urp.2	R	W	88	-7 -		uc002urp.2	R 88 W	possibly damaging	18.58	0	0.93948436
chr1	155583295	155583297	T	SNP	C	nonsynonym	MSTO1		140 exonic	MSTO1:uc031ppy.1:exon11:c.640T>C:p.C214R,MSTO1:uc0	uc001fkw.2	C	R	443	-3	rs112258960	uc001fkw.2	C 443 R	probably damaging	10.43	0	0.12132796
chr9	2181831	2181833	C	SNP	T	nonsynonym	MTAP		123 exonic	MTAP:uc033tzc.1:exon2:c.278C>T:p.T93I,MTAP:uc001ne	uc001nak.1	T	I	93	-6 -		uc001nak.1	T 110 I	probably damaging	25.9	0	0.96039128
chr4	100527931	100527933	C	SNP	A	nonsynonym	MTTP		124 exonic	MTTP:uc003hvc.4:exon12:c.1372C>A:p.L458M,MTTP:uc0	uc003hvc.3	L	M	458	-3 -		uc003hvc.3	L 458 M	probably damaging	16.38	0	0.95097309
chr9	111457804	111457806	C	SNP	T	nonsynonym	MUSK		64 exonic	MUSK:uc022bbs.1:exon4:c.481C>T:p.L161F,MUSK:uc022	uc004ewz.1	L	F	161	-4 -		uc004ewz.1	L 161 F	probably damaging	17.32	0	0.23372352
chr19	50785053	50785053	C	SNP	T	nonsynonym	MYH14		53 exonic	MYH14:uc002prr.1:exon31:c.4369G>T:p.R1457W,MYH14:u	uc002prr.1	R	W	1465	-3 -		uc002prr.1	R 1457 W	probably damaging	17.58	1.00E-04	0.82750779
chr17	10426936	10426938	G	SNP	A	nonsynonym	MYH2		253 exonic	MYH2:uc010cjn.3:exon37:c.5348C>T:p.A1783V,MYH2:uc0	uc002qmp.3	A	V	1783	-5 -		uc002qmp.3	A 1783 V	possibly damaging	28.7	1.00E-04	0.97586399
chr14	23886122	23886124	A	SNP	G	nonsynonym	MYH7		272 exonic	MYH7:uc001wcq.3:exon31:c.4598T>C:p.I1533P,	uc001wcq.2	I	P	1533	-9 -		uc001wcq.2	I 1533 P	probably damaging	23.7	0	0.9942199
chr14	23891397	23891399	C	SNP	T	nonsynonym	MYH7		114 exonic	MYH7:uc001wcq.3:exon25:c.3236G>A:p.R1079Q,	uc001wcq.2	Q	R	1079	5 -		uc001wcq.2	R 1079 Q	benign	16.44	0	0.69724393
chr14	23900798	23900800	G	SNP	A	nonsynonym	MYH7		140 exonic	MYH7:uc001wcq.3:exon8:c.727C>T:p.R243C,	uc001wcq.2	R	C	243	-14 -		uc001wcq.2	R 243 C	probably damaging	16.5	0	0.99335629
chr22	26164334	26164336	G	SNP	A	nonsynonym	MYO1B8		127 exonic	MYO1B8:uc003aca.1:exon2:c.95G>A:p.G32D,MYO1B8:uc0	uc010gur.1	G	D	32	-1 -		uc010gur.1	G 151 D	possibly damaging	16.28	0	0.9575752
chr22	26165099	26165101	G	SNP	A	nonsynonym	MYO1B8		232 exonic	MYO1B8:uc003aca.1:exon2:c.860G>A:p.G287D,MYO1B8:u	uc003aca.1	G	D	287	-1 -		uc003aca.1	G 406 D	benign	11.87	0	0.05868305
chr22	26422531	26422533	C	SNP	T	nonsynonym	MYO1B8		140 exonic	MYO1B8:uc003aca.1:exon41:c.6235C>T:p.R2079C,MYO1	uc010gur.1	R	C	2078	-3 -		uc010gur.1	R 2198 C	benign	10.47	1.00E-04	0.05199946
chr10	68895269	68895271	G	SNP	A	splicing	MYPN		40 splicing	-	-	-	-	-	-30	rs74878633	NaN	NaN	NaN	5.222	0	None
chr10	69918273	69918275	A	SNP	G	nonsynonym	MYPN		77 exonic	MYPN:uc009qzt.3:exon7:c.1349A>G:p.Q450R,MYPN:uc00	uc001jrl.1	Q	R	450	-5 -		uc010qlt.1	Q 450 R	probably damaging	25.7	0	0.96671283
chr5	69405798	69405801	TT	INDEL	T	frameshift_s	NAIP		293 exonic	NAIP:uc003jq.1:exon1:c.445_446A,	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr5	70406475	70406477	G	SNP	C	nonsynonym	NAIP		33 exonic	NAIP:uc003lat.1:exon1:c.122C>G:p.A41G,	uc003lat.1	A	G	41	0 -		uc003lat.1	A 267 G	benign	0.036	0	None
chr5	70406595	70406597	A	SNP	G	nonsynonym	NAIP		51 exonic	NAIP:uc003lat.1:exon1:c.2T>C:p.M1T,	uc003lat.1	M	T	1	-1 -		uc003lat.1	M 227 T	benign	3.936	0	None
chr13	107172206	107172208	G	SNP	A	nonsynonym	NALCN		158 exonic	NALCN:uc001vox.1:exon42:c.4868C>T:p.T1623M,	uc001vox.1	M	T	1623	-1 -		uc001vox.1	T 1623 M	benign	3.958	1.00E-04	0.03022668
chr13	102030955	102030957	T	INDEL	TT	frameshift_i	NALCN		58 exonic	NALCN:uc001vox.1:exon4:c.340_340delinsAA,NALCN:uc0	-	-	-	-	-30 -		NaN	NaN	NaN	NaN	0	None
chr8	134251180	134251182	G	SNP	A	nonsynonym	NDRG1		140 exonic	NDRG1:uc003yue.2:exon3:c.113C>T:p.P38L,	uc003yuq.2	T	T	375	0 -		NaN	NaN	NaN	10.49	0	None
chr2	152347010	152347012	T	SNP	C	nonsynonym	NEB		101 exonic	NEB:uc002tzr.3:exon82:c.8723G>A:p.R2908Q,NEB:uc002	uc002tzr.2	R	Q	2865	1 -		uc002tzr.2	R 304 Q	probably damaging	20.3	1.00E-04	0.21655652
chr2	152348610	152348612	C	SNP	T	splicing	NEB		141 splicing	uc010tbz.2:exon9:c.796+1G>A,uc002tzq.3:exon12:c.112-	-	-	-	-	-30 -		NaN	NaN	NaN	17.62	0	None
chr2	152424651	152424653	C	SNP	A	nonsynonym	NEB		140 exonic	NEB:uc002tzr.3:exon13:c.2072G>T:p.G691V,NEB:uc002t	uc002tzr.2	G	V	648	-3 -		uc010fnx.2	G 4225 V	probably damaging	25.6	0	0.99079508
chr2	152477475	152477477	A	SNP	T	nonsynonym	NEB		87 exonic	NEB:uc002tzu.3:exon72:c.10517T>A:p.I3506N,NEB:uc00	uc010fnx.2	I	N	3251	-3 -		uc010fnx.2	I 3283 N	probably damaging	28.4	0	0.96079576
chr2	152521939	152521941	C	SNP	A	nonsynonym	NEB		167 exonic	NEB:uc002tzu.3:exon42:c.5145G>T:p.K1715N,NEB:uc00	uc010fnx.2	K	N	1715	0 -		uc010fnx.2	K 1715 N	benign	17.32	0	0.65901142
chr2	152522687	152522693	GTAGT	INDEL	GT	frameshift_d	NEB		107 exonic	NEB:uc002tzu.3:exon41:c.4943_4947AC,NEB:uc002tvb.1	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr2	152566277	152566279	C	SNP	T	splicing	NEB		98 splicing	uc022ofb.1:exon11:c.928-1G>A,uc013tpp.1:exon13:c.928-	-	-	-	-	-30 -		NaN	NaN	NaN	23.6	0	None
chr2	152584315	152584346	TGCTGGCTG	INDEL	TGCTGGCTG	nonframeshi	NEB		167 exonic	NEB:uc002tzu.3:exon5:c.154_183CTGGCAGCAGCCAGCA,h	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr22	29884974	29884976	A	SNP	G	nonsynonym	NEFH		140 exonic	NEFH:uc003afo.2:exon4:c.1346A>G:p.K449R,	uc003afo.2	K	R	449	2 -		uc003afo.2	K 449 R	possibly damaging	16.34	1.00E-04	None
chr22	29885127	29885135	AAACAAA	INDEL	AAACAAAAAA	nonframeshi	NEFH		129 exonic	NEFH:uc003afo.3:exon4:c.1489_1505AAACAAAAAACAAA,h	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr22	29885567	29885569	AAGTCCCTC	INDEL	AAGTCCCTC	nonframeshi	NEFH		553 exonic	NEFH:uc003afo.3:exon4:c.1939_1946AAGTCCCTCTGAGAA,h	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr22	29885598	29885608	AGGAAGAGG	INDEL	AGG	nonframeshi	NEFH		42 exonic	NEFH:uc003afo.3:exon4:c.1970_1978AGG,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr22	29885996	29885998	AAG	INDEL	''	nonframeshi	NEFH		0 exonic	NEFH:uc003afo.3:exon4:c.2368_2370del:p.790_790del,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr8	24810373	24810383	CTCCTCCTC	INDEL	CTCCTC	nonframeshi	NEFL		237 exonic	NEFL:uc003exe.4:exon5:c.1573_1583GAGGAG,	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr8	24811096	24811098	G	SNP	A	nonsynonym	NEFL		119 exonic	NEFL:uc003exe.4:exon3:c.1382C>T:p.A461V,	uc003exe.2	A	V	461	0 -		uc003exe.2	A 461 V	benign	7.246	0	None
chr8	24811105	24811107	A	SNP	G	nonsynonym	NEFL		134 exonic	NEFL:uc003exe.4:exon3:c.1373T>C:p.I458T,	uc003exe.2	I	T	458	-1 -		uc003exe.2	I 458 T	benign	14.45	1.00E-04	None
chr15	23086364	23086391	GCCGCCGCCG	INDEL	GCCGCCGCCG	nonframeshi	NIPA1		201 exonic	NIPA1:uc001yxc.3:exon1:c.22_47GCCGCCGCCGCCGCCGCC-	-	-	-	-	301 -		NaN	NaN	NaN	NaN	0	None
chr3	9792057	9792059	T	SNP	C	nonsynonym	OGG1		146 exonic	OGG1:uc003bss.3:exon1:c.88G>T:p.R30C,OGG1:uc003bss.	uc003bss.2	R	C	30	-5		uc003bss.2	R 30 C	probably damaging	16.66	0	0.98134226
chr3	9807702	9807704	C	SNP	T	nonsynonym	OGG1		229 exonic	OGG1:uc003bsm.3:exon7:c.1159C>T:p.H387Y,OGG1:uc003	uc003bsm.2	H	Y	320	2 -		uc003bsm.2	H 387 Y	benign	9.331	0	0.35968819
chr3	193361162	193361164	C	SNP	A	nonsynonym	OPA1		100 exonic;splice	OPA1:uc003lfj.3:exon13:c.1196C>A:p.T399N,OPA1:uc00	uc003lfm.2	T	N	381	-2 -		uc003lfm.2	T 381 N	probably damaging	27.8	0	0.98729903
chr7	92123893	92123895	T	SNP	C	nonsynonym	PEX1		86 exonic

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Chr	LeftFlank	RightFlank	ref_allele	mutltype	var_allele	type	Gene_name	Qual	loc_type	consequence	transcript	ref_aa	var_aa	aa_pos	CDPred_scores	dbID	Polyphen_ts	Polyphen_an	Polyphen_an	CADD_Score	EXAC_POPM	ClinPred
chr19	40900536	40900538	C	SNP	T	nonsynonym	PRX		140 exonic	PRX:uc002orn:3:exon7:c.3722G>A,p.G1241D,PRX:uc002	uc002orn.2	G	D	1102	-1 -		ucl002orn.2	G 1241 D	probably dan	13.96	1.00E-04	0.55556786
chr19	40901322	40901324	C	SNP	T	nonsynonym	PRX		118 exonic	PRX:uc002orn:3:exon7:c.2936G>A,p.G979E,PRX:uc002or	uc002orn.2	G	E	979	-2 -		ucl002orn.2	G 979 E	possibly dam	8.682	0	0.91279781
chr19	40901679	40901681	T	SNP	G	nonsynonym	PRX		62 exonic	PRX:uc002orn:3:exon7:c.2579A>C,p.D860A,PRX:uc002or	uc002orn.2	D	A	721	-2 -		ucl002orn.2	D 860 A	possibly dam	9.075	0	0.48988537
chr6	32810784	32810786	C	SNP	T	nonsynonym	PSMB8		140 exonic	PSMB8:uc003oce.3:exon2:c.229G>A:p.A77T,PSMB8:uc00	uc003oce.2	A	T	73	-7 -		ucl003oce.2	A 77 T	probably dan	36	0	0.97664273
chr11	64514803	64514805	C	SNP	T	nonsynonym	PYGM		71 exonic	PYGM:uc001oax.4:exon18:c.2204G>A:p.R735H,PYGM:uc	uc001oax.3	R	H	735	2 -		ucl001oax.3	R 735 H	benign	15.42	1.00E-04	0.65858871
chr7	127976096	127976098	C	SNP	T	splicing	RBM28		193 splicing	uc003nmp.2:exon6:c.614-1G>A,uc003ksq.1:exon4:c.191-	-	-	-		-30 -		NaN	NaN	NaN	12.77	0	None
chr2	161223876	161223878	A	SNP	T	nonsynonym	RBMS1		86 exonic	RBMS1:uc002ubm.3:exon3:c.27>A:p.M1K,RBMS1:uc002ubl.2		M	K	32	-1 -		ucl002ubm.2	M 34 K	benign	22.4	0	0.94663972
chr8	101271358	101271360	C	SNP	T	nonsynonym	RNF19A		112 exonic	RNF19A:uc003ygk.2:exon10:c.1942G>A:p.G648S,RNF19A	uc003ygk.1	G	S	648	0 -		ucl003ygk.1	G 648 S	benign	15.05	1.00E-04	0.06997599
chr11	124750447	124750461	CGGAGTCGG	INDEL	CGGAGTC	nonframeshi	ROBO3		915 exonic	ROBO3:uc001qbd.2:exon11:c.868_880DGGAGTC,ROBO3	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr8	101250839	101250841	G	INDEL	GG	frameshift_s	RPM28		117 exonic	RPM28:uc012aad.1:exon1:c.211_211delinsCC,	-	-	-		-10 -		NaN	NaN	NaN	NA	0	None
chr19	38945967	38945969	G	SNP	A	nonsynonym	RYR1		257 exonic	RYR1:uc002ozu.3:exon14:c.1534G>A:p.E512K,RYR1:uc00	uc002ozu.2	E	K	512	5 -		ucl002ozu.2	E 512 K	probably dan	15.95	0	0.96318388
chr19	38948770	38948772	A	SNP	G	nonsynonym	RYR1		140 exonic	RYR1:uc002ozu.3:exon18:c.2006A>G:p.E669G,RYR1:uc00	uc002ozt.2	E	G	669	1 -		ucl002ozt.2	E 669 G	possibly dam	14.07	0	0.88879639
chr19	38960073	38960075	T	SNP	C	nonsynonym	RYR1		223 exonic	RYR1:uc002ozu.3:exon27:c.3608T>C,p.M1229T,RYR1:uc0	uc002ozt.2	M	T	1229	-1 -		ucl002ozt.2	M 1229 T	possibly dam	10.31	0	0.88757968
chr19	38966090	38966092	T	SNP	C	nonsynonym	RYR1		109 exonic	RYR1:uc002ozu.3:exon31:c.4471T>C,p.C1491R,RYR1:uc00	uc002ozu.2	C	R	1491	-10 -		ucl002ozt.2	C 1491 R	probably dan	14.83	0	0.99895275
chr13	23911312	23911314	A	SNP	T	nonsynonym	SAC5		103 exonic	SAC5:uc001uao.3:exon10:c.670T>A:p.F223L,SAC5:uc00	uc001uao.2	F	L	2087	0 -		ucl001uao.2	F 223 L	benign	14.95	0	0.30140662
chr13	23928729	23928731	C	SNP	T	nonsynonym	SAC5		107 exonic	SAC5:uc001uao.1:exon1:c.1382G>A:p.G461D,SAC5:uc00	uc001uao.2	G	D	527	-1 -		ucl001uao.1	G 461 D	probably dan	14.94	0	0.84409225
chr22	50900428	50900430	G	SNP	A	nonsynonym	SBF1		140 exonic	SBF1:uc003bih.2:exon20:c.2519C>T:p.T840M,SBF1:uc00	uc003bih.2	M	T	839	-1 -		ucl003bih.2	T 839 M	probably dan	14.89	1.00E-04	None
chr22	50903538	50903540	A	SNP	T	nonsynonym	SBF1		140 exonic	SBF1:uc003bih.2:exon12:c.1236G>A:p.M409I,SBF1:uc00	uc003bih.2	H	H	409	-8 -		ucl003bih.2	H 408 H	probably dan	27.8	0	None
chr22	50913214	50913216	C	SNP	G	nonsynonym	SBF1		72 exonic,splice	SBF1:uc003bih.2:exon1:c.55G>C:p.G19R,SBF1:uc003bih.	uc003bih.2	G	R	19	0 -		ucl003bih.2	G 19 R	possibly dam	12.68	0	None
chr11	9861054	9861056	G	SNP	C	nonsynonym	SBF2		86 exonic	SBF2:uc001mdl.3:exon25:c.2713C>G:p.L905V,SBF2:uc00	uc001mdl.2	L	V	1149	-1 -		ucl001mdl.2	L 1149 V	benign	18.04	0	0.29172274
chr2	167262288	167262290	G	SNP	T	nonsynonym	SCN7A		191 exonic	SCN7A:uc002udu.2:exon25:c.4850C>A:p.T1617K,	uc002udu.1	T	K	1617	-1 -		ucl002udu.1	T 1617 K	probably dan	18.07	0	0.98859006
chr2	167266239	167266241	C	SNP	T	nonsynonym	SCN7A		105 exonic	SCN7A:uc002udu.2:exon24:c.3917G>A:p.R1306H,	uc002udu.1	R	H	1306	-3 -		ucl002udu.1	R 1306 H	benign	10.68	0	0.03969857
chr17	10584501	10584503	G	SNP	C	nonsynonym	SCO1		114 exonic	SCO1:uc002gnc.4:exon6:c.840C>G:p.N280K,	uc002gnc.3	N	K	249	-3 -		ucl002gnc.3	N 249 K	possibly dam	27.9	0	0.98114145
chr17	75484869	75484871	A	SNP	G	nonsynonym	SEPT9		83 exonic	SEPT9:uc002jty.4:exon5:c.413A>G:p.N145D,SEPT9:uc01	uc002jty.2	N	D	389	-5 -		ucl002jty.2	N 145 D	possibly dam	17.87	0	0.81069171
chr9	135203528	135203530	A	SNP	C	nonsynonym	SETX		230 exonic	SETX:uc010nxr.3:exon11:c.2313T>G:p.F1771,SETX:uc00	uc010nxr.2	F	L	1152	0 -		ucl010nxr.2	F 1152 L	benign	9.514	0	0.07288013
chr9	135203910	135203929	TCATCATCAT	INDEL	TCATCATCAT	nonframeshi	SETX		114 exonic	SETX:uc010nxr.3:exon1:c.1914_1931TGATGATGATGATGAT	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr9	135204769	135204771	C	SNP	T	nonsynonym	SETX		138 exonic	SETX:uc010nxr.3:exon1:c.1072G>A:p.G358R,SETX:uc00	uc010nxr.2	G	R	739	2 -		ucl010nxr.2	G 739 R	possibly dam	18.58	0	0.87888773
chr5	156184725	156184731	AGAAG	INDEL	AG	nonframeshi	SGCD		241 exonic	SGCD:uc003wxb.3:exon8:c.710_714AG,	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr13	23771285	23771287	G	SNP	A	splicing	SGCG		91 splicing	-	-	-	-		-30	rs115717101	NaN	NaN	NaN	3.111	0	None
chr5	148392231	148392233	G	SNP	C	nonsynonym	SHSTC2		179 exonic	SHSTC2:uc010gpw.3:exon4:c.205G>C:p.T68R,SHSTC2:uc010gpw.2		T	R	1033	-1 -		ucl010gpw.2	T 1033 R	benign	2.364	1.00E-04	0.01975677
chr5	148406434	148406436	G	SNP	A	stopgain_SN	SHSTC2		140 exonic	SHSTC2:uc010gpw.3:exon12:c.1501C>T:p.R501X,SHSTC2:u	uc010dbz.1	R	*	839	-30 -		NaN	NaN	NaN	37	0	None
chr5	148407255	148407257	T	SNP	C	nonsynonym	SHSTC2		140 exonic	SHSTC2:uc010gpw.3:exon12:c.680A>G:p.Y227C,SHSTC2:u	uc010gpw.2	Y	C	324	-2 -		ucl010gpw.2	Y 680 C	probably dan	17.3	1.00E-04	0.99654547
chr5	148418028	148418030	G	SNP	A	nonsynonym	SHSTC2		112 exonic	SHSTC2:uc003ipu.3:exon8:c.830C>T:p.T277M,SHSTC2:uc	uc011dbz.1	T	M	162	-1 -		ucl011dbz.1	T 162 M	benign	12.01	1.00E-04	0.04260866
chr5	148422280	148422282	A	SNP	G	nonsynonym	SHSTC2		92 exonic	SHSTC2:uc003ipu.3:exon5:c.505T>C:p.Y169H,SHSTC2:uc0	uc003ipu.2	Y	H	169	2 -		ucl003ipu.2	Y 169 H	possibly dam	13.76	0	0.04089892
chr17	73269538	73269540	T	SNP	C	nonsynonym	SLC25A19		140 exonic	SLC25A19:uc002ov.4:exon8:c.956A>G:p.Q319R,SLC25A1	uc002ov.3	Q	R	319	1 -		ucl002ov.3	Q 262 R	benign	4.827	0	0.02624725
chr20	746113	746115	C	SNP	T	nonsynonym	SLC52A3		153 exonic	SLC52A3:uc002wwd.4:exon2:c.305G>A:p.G102D,SLC52A	uc002wwe.3	G	D	102	-1 -		ucl002wwd.3	G 102 D	benign	0.088	0	0.10315216
chr2	108626991	108626993	A	SNP	G	nonsynonym	SLC5A7		240 exonic	SLC5A7:uc010fbj.3:exon9:c.1418A>G:p.Y473C,SLC5A7:uc	uc010pwm.1	Y	C	360	-2 -		ucl010pwm.1	Y 473 C	possibly dam	9.122	0	0.55472201
chr2	220347867	220347869	G	SNP	A	nonsynonym	SPEG		93 exonic	SPEG:uc010fwg.3:exon30:c.5683G>A:p.E1895K,	uc010fwg.2	E	K	1895	1 -		ucl010fwg.2	E 1895 K	probably dam	16.09	0	0.9213565
chr15	44888343	44888345	C	SNP	G	nonsynonym	SPG11		231 exonic	SPG11:uc010aeh.2:exon25:c.4371G>C:p.W1457C,SPG11	uc001etx.2	W	C	1457	-2 -		ucl010aeh.1	W 1457 C	possibly dam	16.31	0	0.67337423
chr15	44907653	44907655	G	SNP	A	nonsynonym	SPG11		141 exonic	SPG11:uc010aeh.2:exon16:c.2945C>T:p.T982I,SPG11:uc	uc001etx.2	T	I	982	-1 -		ucl001etx.2	T 982 I	benign	14.26	0	0.18115255
chr15	44912608	44912617	AAAAAAAAA	INDEL	AAAAAAAAA	frameshift_s	SPG11		66 exonic	SPG11:uc001etz.1:exon1:c.83_90TTTTTTTT,	-	-	-		-30 -		NaN	NaN	NaN	NA	0	None
chr13	36888362	36888364	C	SNP	A	nonsynonym	SPG20		97 exonic	SPG20:uc001uxm.3:exon6:c.1852C>T:p.T551I,SPG20:uc0	uc001uxm.2	T	I	551	-2 -		ucl001uxm.1	T 551 I	possibly dam	23.3	0	0.92013627
chr16	89576939	89576941	A	SNP	G	nonsynonym	SPG7		88 exonic	SPG7:uc002fri.3:exon2:c.226A>G:p.N76D,SPG7:uc002fri	uc002fri.2	N	D	76	1 -		ucl002fri.2	N 76 D	benign	5.754	0	0.02401603
chr16	89597198	89597200	T	SNP	A	nonsynonym	SPG7		112 exonic	SPG7:uc002fri.3:exon7:c.970T>A:p.F324,SPG7:uc002fri	uc002fri.2	F	I	324	5 -		ucl002fri.2	F 324 I	probably dan	24.4	0	0.97971218
chr16	89615453	89615455	G	SNP	A	nonsynonym	SPG7		80 exonic	SPG7:uc002fri.4:exon7:c.1540>A:p.D52N,	uc002fri.2	N	N	52	1 -		NaN	NaN	NaN	3.501	0	0.06612203
chr19	41062929	41062931	T	SNP	G	nonsynonym	SPTBN4		25 exonic,splice	SPTBN4:uc002omy.3:exon26:c.5291T>G:p.V1764G,SPTBN	uc002omy.2	V	G	1764	-1 -		ucl002omy.2	V 507 G	probably dam	19.34	0	0.99655801
chr19	41062973	41062975	G	SNP	A	nonsynonym	SPTBN4		175 exonic	SPTBN4:uc002omy.3:exon26:c.5335G>A:p.A1779T,SPTBN	uc002omy.2	T	T	1779	-4 -		ucl002omy.2	T 521 T	possibly dam	10.44	0	0.70026336
chr9	94809515	94809517	G	SNP	A	nonsynonym	SPTLC1		132 exonic	SPTLC1:uc004arl.1:exon11:c.1019C>T:p.S340L,SPTLC1:uc	uc011lhv.1	S	L	340	-8 -		uc004arl.1	S 340 L	probably dan	33	1.00E-04	0.99031758
chr5	179259914	179259916	A	SNP	T	nonsynonym	SQSTM1		238 exonic	SQSTM1:uc003mbs.3:exon2:c.107A>T:p.N36I,SQSTM1:uc	uc003mbw.3	N	I	36	-3 -		uc003mbw.3	N 120 I	benign	16.25	0	0.69377649
chr9	136219586	136219588	T	SNP	C	nonsynonym	SURF1		273 exonic	SURF1:uc004cqb.1:exon6:c.550A>G:p.R184G,	uc004cqb.1	R	G	75	-8 -		uc004cqb.1	R 184 G	possibly dam	16.71	0	0.98891661
chr3	12228860	12228864	TCTTCCTCCTI	INDEL	TCTTCCTCCTI	nonframeshi	SYN2		410 exonic	SYN2:uc003bem.3:exon9:c.484_506TCTTCCTCCTCTC,SYN2:u	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr6	152451861	152451863	G	SNP	A	nonsynonym	SYNE1		140 exonic	SYNE1:uc003qau.4:exon145:c.26146C>T:p.H8167Y,SYNE	uc003qau.3	H	Y	3240	2 -		ucl003qau.3	H 8716 Y	benign	16.4	0	0.91388601
chr6	152470754	152470756	T	SNP	C	nonsynonym	SYNE1		116 exonic	SYNE1:uc003qau.4:exon136:c.24499A>G:p.I8167V,SYNE	uc003qau.3	I	V	329	-2 -		ucl003qau.3	I 369 V	benign	15.12	1.00E-04	0.26476732
chr6	152464649	152464651	T	SNP	G	nonsynonym	SYNE1		40 exonic	SYNE1:uc003qau.4:exon82:c.15880A>C:p.M5294L,SYNE1	uc003qau.3	M	L	5294	2 -		ucl003qau.3	M 5223 L	benign	5.211	1.00E-04	0.01382485
chr6	152464762	152464764	G	SNP	T	nonsynonym	SYNE1		60 exonic	SYNE1:uc003qau.4:exon82:c.15767C>A:p.T5256K,SYNE1	uc003qau.3	T	K	5185	-1 -		ucl003qau.3	T 5185 K	benign	2.586	0	0.21388236
chr6	152466393	152466395	G	SNP	A	nonsynonym	SYNE1		90 exonic	SYNE1:uc003qau.4:exon81:c.15482C>T:p.A5161V,SYNE1	uc010kxz.2	A	V	5161	-4 -		ucl010kxz.2	A 5161 V	benign	12.27	0	0.42708623
chr6	152647180	152647182	A	SNP	G	nonsynonym	SYNE1		90 exonic	SYNE1:uc003qau.4:exon80:c.15150T>C:p.M5117T,SYNE1	uc010kxz.2	M	T	5046	-5 -		ucl010kxz.2	M 5117 T	possibly dam	16.61	0	0.46143356
chr6	152652720	152652722	T	SNP	C	nonsynonym	SYNE1		140 exonic	SYNE1:uc003qau.4:exon78:c.13099A>G:p.I4367V,SYNE1	uc003qau.3	I	V	122	3 -		ucl003qau.3	I 4367 V	benign	0.847	0	0.10961259
chr6	152722349	152722351	T	SNP	C	nonsynonym	SYNE1		114 exonic	SYNE1:uc003qau.4:exon47:c.6953A>G:p.T2318A,SYNE1	uc003qau.3	T	A	2325	0 -		ucl003qau.3	T 2325 A	benign	10.1	0	0.08855259
chr6	152823765	152823767	A	SNP	T	splicing	SYNE1		65 splicing	uc003qat.4:exon11:c.909+2T>A,uc003qau.4:exon11:c.88	-	-	-		-30 -		NaN	NaN	NaN		0	None
chr14	64453330	64453332	T	SNP	C	nonsynonym	SYNE2		115 exonic	SYNE2:uc001egl.3:exon19:c.2307T>C:p.L770S,SYNE2:uc0	uc001egl.2	L	S	770	-2 -		ucl001egl.2	L 770 S	probably dan	12.91	0	0.95218813
chr14	64457711	64457713	C	SNP	G	nonsynonym	SYNE2		83 exonic	SYNE2:uc001egl.3:exon21:c.2525C>G:p.P842R,SYNE2:uc	uc001egl.2	P	R	842	-2 -		ucl001egl.2	P 842 R	benign	10.86	0	0.18484244
chr14	64486749	64486751	C	SNP	T	nonsynonym	SYNE2		55 exonic	SYNE2:uc001egl.3:exon35:c.5090C>T:p.S1697F,SYNE2:u	uc001egl.2	S	F	1697	-2 -		ucl001egl.2	S 1697 F	benign	12.75	0	0.41384098
chr14	64514816	64514818	A	SNP	G	nonsynonym	SYNE2		79 exonic	SYNE2:uc001egl.3:exon46:c.7121A>G:p.N2441D,SYNE2:u	uc001egl.2	N	D	2441	1 -		ucl001egl.2	N 2441 D	benign	11.37	0	0.32703424
chr14	64608747	64608749	A	SNP	G	nonsynonym	SYNE2		140 exonic	SYNE2:uc001egn.3:exon3:c.134A>G:p.D45G,SYNE2:uc00	uc001egm.2	D	G	5083	-6 -		uc001egm.2	D 5083 G	benign	6.095	0	0.02043282
chr11	7324356	7324358	G	SNP	T	nonsynonym	SYT9		105 exonic	SYT9:uc001mfe.3:exon2:c.233G>T:p.C78F,	uc001mfe.2	C	F	78	-2 -		ucl001mfe.2	C 78 F	benign	16.19	0	0.39121538
chr17	34171846	34171848	GGAGGAGA	INDEL	GGAGGAGA	nonframeshi	TAF15		501 exonic	TAF15:uc002hk6.4:exon15:c.1504_1544GGAGGAGATCG	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr17	34171894	34171896	A	SNP	G	nonsynonym	TAF15		343 exonic	TAF15:uc002hk6.4:exon15:c.1592A>G:p.Y531C,TAF15:uc	uc002hkc.2	Y	C	528	-2 -		ucl002hkd.2	Y 531 C	benign	12.46	1.00E-04	0.06040257
chr22	20052090	20052092	C	SNP	T	nonsynonym	TANGO2		90 exonic	TANGO2:uc010ayo.2:exon10:c.737C>T:p.A246V,TANGO2	uc010zvf.2	A	V	184	-4 -		ucl010zvf.2	A 248 V	benign	6.811	0	0.06433446
chr6	134210727	134210729	A	SNP	G	nonsynonym	TCF21		101 exonic	TCF21:uc003qei.4:exon1:c.193A>G:p.K65E,TCF21:uc003	uc003qei.3	K	E	65	1 -		ucl003qei.3	K 65 E	benign	16.29	0	0.06721938
chr14	90489419	90489424	TGGT	INDEL	T	frameshift_s	TDP1		140 exonic	TDP1:uc010ans.3:exon10:c.1689_1690T,	-	-	-		-30 -		NaN	NaN	NaN	NA	0	None
chr11	85342836	85342838	A	SNP	G	nonsynonym	TMEM126B		50 exonic	TMEM126B:uc001pao.4:exon1:c.98A>G:p.Y33C,TMEM126	uc001pao.1	Y	C	33	-7 -		uc001pao.1	Y 33 C	benign	2.395	0	0.02222801
chr12	64202605	64202607	G	SNP	A	nonsynonym	TMEM5		92 exonic	TMEM5:uc001ung.2:exon6:c.1006G>A:p.V356M,TMEM5:uc	uc001ung.1	V	M	356	1 -		uc001ung.1	V 356 M	possibly dam	15.83	1.00E-04	0.59951574
chr6	31979405	31979407	G	SNP	A	nonsynonym	TNXB		69 exonic	TNXB:uc021yef.2:exon2:c.448C>T:p.R150W,	uc011dpc.1	R	W	150	-3 -		ucl011dpc.1	R 3746 W	possibly dam	4.061	0	None
chr6	31979461	31979463	C	SNP	T	nonsynonym	TNXB		36 exonic	TNXB:uc021yef.2:exon2:c.392G>A:p.R131H,	uc011dpc.1	R	H	131	0 -		ucl011dpc.1	R 3747 H	probably dam	1.873	0	None
chr6	32010501	32010503	G	SNP	A	nonsynonym	TNXB		69 exonic	TNXB:uc003nzl.2:exon39:c.12026C>T:p.P4009L,TNXB:uc	uc003nzl.2	P	L	478	-3 -		uc003nzl.2	P 4009 L	probably dam	11.81	0	0.86017227
chr6	32029205	32029207	C	SNP	T	nonsynonym	TNXB		82 exonic	TNXB:uc003nzl.2:exon21:c.7460G>A:p.R2487H,	uc003nzl.2	R	H	2487	0 -		uc003nzl.2	R 2487 H	probably dam	14.07	0	0.97874177
chr6	32037476	32037478	C	SNP	T	nonsynonym	TNXB		97 exonic	TNXB:uc003nzl.2:exon15:c.5440G>A:p.D1814N,	uc003nzl.2	D	N	1814	1 -		uc003nzl.2	D 1814 N	possibly dam	17.32	0	0.73719317
chr6	32061680	32061682	C	SNP	T	nonsynonym	TNXB		393 exonic	TNXB:uc003nzl.2:exon9:c.1949G>A:p.R650H,	uc003nzl.2	R	H	650	0	rs 17201660	uc003nzl.2	R 650 H	probably dam	11	0	0.02223468
chr4	184606528	184606530	A	SNP	G	nonsynonym	TRAPPC11		125 exonic	TRAPPC11:uc003hnw.3:exon17:c.1735A>G:p.I579V,TRAP	uc003hnw.2	I	V	185	3 -		uc003hnw.2	I 579 V	benign	13.78	0	0.44373915
chr6	123786032	123786045	AAAAAAAAA	INDEL	AAAAAAAAA	frameshift_s	TRDN		156 exonic	TRDN:uc010kwn.3:exon9:c.880_889TTTTTTTTT,	-	-	-		-30 -		NaN	NaN	NaN	NA	0	None
chr6	123837298	123837312	TTTTTCTTTT	INDEL	TTTTTCT	nonframeshi	TRDN		48 exonic	TRDN:uc010kwn.3:exon6:c.525_537AGAAAAAA,TRDN:uc0	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr4	154191576	154191578	C	SNP	T	nonsynonym	TRIM2		279 exonic	TRIM2:uc003ln.1:exon2:c.94C>T:p.R32C,TRIM2:uc003lnf	uc003lnh.2	R	C	41	-4 -		uc003lng.2	R 14 C	probably dan	31	1.00E-04	0.76335657
chr15	64701884	64701886	A	SNP	G	nonsynonym	TRIP4		138 exonic	TRIP4:uc002xmn.3:exon7:c.889A>G:p.K300R,	uc002xmn.2	K	R	300	2 -		ucl002xmn.2	K 300 R	benign	22.4	1.00E-04	0.152957
chr12	110224530	110224532	G	SNP	A	nonsynonym	TRPV4		141 exonic	TRPV4:uc001tpk.2:exon14:c.2320C>T:p.R774C,TRPV4:uc	uc001tpk.1	R	C	774	-3 -		uc001tpj.1	R 774 C	possibly dam	17.49	1.00E-04	0.78040302
chr12	110252564	110252566	C	SNP	A	nonsynonym	TRPV4		31 exonic	TRPV4:uc001tpk.2:exon2:c.370>A:p.G138,TRPV4:uc012	uc001tpk.1	G	R	13	-2 -		uc001tpj.1	G 13 R	probably dam	11.55	1.00E-04	0.092179
chr2	179404205	179404207	C	SNP	A	nonsynonym	TTN		194 exonic	TTN:uc021vfa.2:exon181:c.71766G>T:p.E23922D,TTN:uc	uc010ufr.1	E	D	23918	-5 -		ucl010ufn.1	E 23989 D	probably dam	13.07	0	0.3826479
chr2	179404267	179404269	C	SNP	A	nonsynonym	TTN		356 exonic	TTN:uc021vfa.2:exon181:c.71704G>T:p.A23902S,TTN:uc	uc010ufr.1	A	S	23773	1 -		ucl010ufr.1	A 23902 S	possibly dam	22.6	0	0.41646316
chr2	179404836	179404838	G	SNP	T	nonsynonym	TTN		226 exonic	TTN:uc021vfa.2:exon180:c.71236C>A:p.P23746T,TTN:uc	uc010ufr.1	P	T	23742	-1 -		ucl010ufq.1	P 23045 T	probably dam	15.5	0	0.45078269
chr2	179413173	179413175	C	SNP	T	nonsynonym	TTN		334 exonic	TTN:uc021vfa.2:exon168:c.66359G>A:p.R22132H,TTN:uc	uc010ufj.1	R	H	21991	3 -		ucl010ufj.1	R 21995 H	probably dam	21	1.00E-04	0.61228377
chr2	179416433	179416435	C	SNP	T	nonsynonym	TTN		246 exonic	TTN:uc021vfa.2:exon164:c.64373G>A:p.R21458H,TTN:uc	uc010ufi.1	R	H	21454	-6 -		ucl010ufi.1	R 21525 H	possibly dam	17.61	1.00E-04	0.89600137
chr2	179425012	179425014	G	SNP	A	nonsynonym	TTN		245 exonic	TTN:uc021vfa.2:exon155:c.59026C>T:p.R19676W,TTN:u	uc010ufg.1	R	W	26040	-7 -		ucl010ufi.1	R 19676 W	possibly dam	11.2	0	0.64891505
chr2	179426556	179426558	G	SNP	A	nonsynonym	TTN		184 exonic	TTN:uc021vfa.2:exon155:c.57482T>T:p.T19161,TTN:uc	uc010ufi.1	T	I	19157	-4 -		ucl010ufg.1	T 26460 I	benign	11.25	0	0.37542414
chr2	179429187	179429189	T	SNP	C	nonsynonym	TTN		201 exonic	TTN:uc021vfa.2:exon155:c.54853A>G:p.N18284S,TTN:u	uc010ufi.1	N	S	18280	3 -		ucl010ufi.1	N 18159 S	probably dam	11.38	0	0.12695584
chr2	179437756	179437758	C	SNP	A	nonsynonym	TTN		144 exonic	TTN:uc021vfa.2:exon155:c.46282G>T:p.D15428Y,TTN:u	uc010ufi.1	D	Y	15299	-6 -		ucl010ufi.1	D 15428 Y	possibly dam	13.5	0	0.64708439
chr2	179437785	179437787	T	SNP	C	nonsynonym	TTN		140 exonic	TTN:uc021vfa.2:exon155:c.46253A>G:p.Y15418C,TTN:uc	uc010ufi.1	Y	C	15414	-14 -		uc010ufi.1	Y 15485 C	probably dam	12.03	1.00E-04	0.39448243
chr2	179438142	179438144	A	SNP	T	nonsynonym	TTN		195 exonic	TTN:uc021vfa.2:exon155:c.45896T>A:p.M15299K,TTN:uc	uc010ufi.1	M	K	15295	-9 -		ucl010ufj.1	M 15174 K	benign	11.59	0	0.32839394
chr2	179439083	179439085	A	SNP	C	nonsynonym	TTN		348 exonic	TTN:uc021vfa.2:exon155:c.44955T>G:p.U14985M,TTN:uc	uc010ufi.1	M	M	15044	1 -		ucl010ufi.1	I 14985 M	possibly dam	2.187	0	0.1282583
chr2	179446426	179446428	C	SNP	G	nonsynonym	TTN		330 exonic	TTN:uc021vfa.2:exon142:c.39748G>C:p.G13250R,TTN:uc	uc010ufh.1	G	R	13246	-4 -		uc010ufh.1	G 13125 R	possibly dam	17.26	0	0.77339423
chr2	179447684	179447686	T	SNP	C	nonsynonym	TTN		129 exonic	TTN:uc021vfa.2:exon142:c.39025A>G:p.I13009V,TTN:uc	uc010ufh.1	I	V	12880	1 -		uc010ufh.1	I 13076 V	benign	14.64	0	0.24567722
chr2	179455397	179455399	C	SNP	T	nonsynonym	TTN		144 exonic	TTN:uc021vfa.2:exon131:c.34234G>A:p.G11412R,TTN:u	uc010ufg.1	G	R	11408	-7 -		uc010ufg.1	G 10711 R	possibly dam	13.57	0	0.56014591
chr2	179460427	179460429	T	SNP	A	nonsynonym	TTN		161 exonic	TTN:uc021vfa.2:exon124:c.30833A>T:p.N10278I,TTN:uc	uc010ufg.1	N	I	16642	-5 -		uc010ufg.1	N 10153 I	possibly dam	17.33	0	0.89431769
chr2	179479224	179479226	T	SNP	C	nonsynonym	TTN		252 exonic	TTN:uc021vfa.2:exon90:c.22196G>A:p.R7399Q,TTN:uc0	uc010ufe.1	R	Q	7399	4 -		uc010ufg.1	R 14658 Q	possibly dam	17.39	0	0.51268456
chr2	179511767	179511790	TATATATATA	INDEL	TATATATATA	splicing	TTN		171 splicing	-	-	-	-		-30 -		NaN	NaN	NaN	NA	0	None
chr2	179514542	179514544	C	SNP	A	splicing	TTN		116 splicing	uc003rgc.1:exon212:c.39895+1G>T,uc003rgd.1:exon167:-	-	-	-		-30 -		NaN	NaN	NaN	19.24	0	None
chr2	179514941	179514985	TTTCCTTCTG	INDEL	TTTCCTTCTG	nonframeshi	TTN		93 exonic	TTN:uc003rgd.1:exon165:c.35203_35245GAGGAGAAATTGG	-	-	-		301 -		NaN	NaN	NaN	NA	0	None
chr2	179530115	179530117	G	SNP	A	nonsynonym	TTN		92 exonic	TTN:uc010ufk.