Table S5. All variants with annotations and frequencies.

Gene	Chromosome	Position	Reference Allele	Alternate Allele	Known Variation	Truncating	CADD Consequence Detail	HGVS CDS Notation	HGVS Protein Notation	Amino Acid Substitution	Protein Domain	CADD Scaled Score	PolyPhen2 Category	PolyPhen2 Score	SIFT Category	SIFT Score	AlignGVGD Score	ExAC Frequency	Non-Finnish European Carrier Frequency
ATM	11	108089353	T	C	CM960095,COSM1501141,COSM1561140	NO	initiator_codon	ENST00000276616.4:c.27:C>	ENSP00000276616.4:p.Met1?	M1T	PTIR11139.PTIR11139:SF166	25	probably_damaging	0.997	deleterious	0	NA	0.00000	
ATM	11	108089354	G	A	CM059187,rs781404312	NO	initiator_codon	ENST00000276616.4:c.2C>A	ENSP00000276616.4:p.Met1?	M1I	PTIR11139.PTIR11139:SF166	37	probably_damaging	0.995	deleterious	0	NA	0.00000	
ATM	11	108089386	G	A	NA	NO	missense	ENST00000276616.4:c.35G>A	ENSP00000276616.4:p.Cys12Tyr	C12Y	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	26.6	probably_damaging	1	deleterious	0	65	0.00000	
ATM	11	108089389	G	A	rs778201041,COSM1183947,COSM1183946	NO	missense	ENST00000276616.4:c.38G>A	ENSP00000276616.4:p.Arg13His	R13H	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	22.8	benign	0.176	tolerated	0.21	0	0.00006	
ATM	11	108089394	T	C	NA	NO	synonymous	ENST00000276616.4:c.39T:C	NA	R13R	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	4.066	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089393	AC	A	rs771887195	YES	frameshift_feature_truncation	ENST00000276616.4:c.43dAC	ENSP00000276616.4:p.Leu15Ter	L15X	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	25.6	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089407	G	A	NA	NO	missense	ENST00000276616.4:c.56G>A	ENSP00000276616.4:p.Arg19Lys	R19K	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	9.874	benign	0.012	tolerated	1	0	0.00006	
ATM	11	108089417	A	G	NA	NO	synonymous	ENST00000276616.4:c.66A>G	NA	E22E	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	8.13	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089443	A	T	rs761335619	NO	synonymous	ENST00000276616.4:c.92-22A>T	NA	NA	NA	3.755	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089501	A	G	CM051444	YES	splice_acceptor	ENST00000276616.4:c.73-2A>G	NA	NA	NA	24.9	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089504	A	G	NA	NO	missense	ENST00000276616.4:c.76A>T	ENSP00000276616.4:p.Glu26Asp	E26D	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	23.5	possibly_damaging	0.528	tolerated	0.12	0	0.00006	
ATM	11	108089514	G	A	NA	NO	synonymous	ENST00000276616.4:c.86G>A	NA	E29E	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	11.65	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089538	C	T	rs2218684	NO	missense	ENST00000276616.4:c.113C>T	ENSP00000276616.4:p.Arg45Trp	R45W	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	28.9	possibly_damaging	0.893	deleterious	0	25	0.00006	
ATM	11	108089568	T	C	CD982445,rs770834907	NO	synonymous	ENST00000276616.4:c.138T:C	NA	H46H	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	0.142	NA	NA	NA	NA	NA	0.00003	
ATM	11	108089576	C	G	CM990201,rs1800054	NO	missense	ENST00000276616.4:c.146C>G	ENSP00000276616.4:p.Ser49Cys	S49C	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	25.1	possibly_damaging	0.841	deleterious	0.01	15	0.02247	
ATM	11	108089592	T	C	rs2218690	NO	synonymous	ENST00000276616.4:c.162T:C	NA	Y54Y	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	0.051	NA	NA	NA	NA	NA	0.00498	
ATM	11	108089598	T	G	NA	NO	missense	ENST00000276616.4:c.167T>G	ENSP00000276616.4:p.Asn56Lys	N56K	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	26.3	probably_damaging	0.93	deleterious	0.04	15	0.00000	
ATM	11	108089606	C	T	rs786880388	NO	missense	ENST00000276616.4:c.176C>T	ENSP00000276616.4:p.Ala59Val	A59V	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	26.3	possibly_damaging	0.826	tolerated	0.05	0	0.00000	
ATM	11	108089614	A	G	NA	NO	synonymous_splice	ENST00000276616.4:c.184A>G	ENSP00000276616.4:p.Arg62Gly	R62G	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	26.9	probably_damaging	0.938	deleterious	0.01	0	0.00000	
ATM	11	108089623	A	G	rs753891198,COSM3970880,COSM3979679	NO	splice_intron	ENST00000276616.4:c.185+8A>G	NA	NA	NA	9.836	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089640	T	A	NA	NO	intron	ENST00000276616.4:c.185+253T>A	NA	NA	NA	13.54	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089655	C	CTA	NA	NO	intron	ENST00000276616.4:c.185+41_185+42insAT	NA	NA	NA	8.097	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089660	A	G	rs759014058	NO	intron	ENST00000276616.4:c.185+45A>G	NA	NA	NA	2.911	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089664	G	A	NA	NO	intron	ENST00000276616.4:c.186-617>G	NA	NA	NA	1.023	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089659	TTAAG	T	rs778876770	NO	intron	ENST00000276616.4:c.186-43_186-40delAGTA	NA	NA	NA	8.784	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089670	A	G	NA	NO	intron	ENST00000276616.4:c.186-35A>G	NA	NA	NA	11.34	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089671	C	T	rs371745892	NO	intron	ENST00000276616.4:c.186-34C>T	NA	NA	NA	10.83	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089672	G	A	rs771729439	NO	intron	ENST00000276616.4:c.186-33G>A	NA	NA	NA	11.91	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089686	G	A	rs748791429	NO	intron	ENST00000276616.4:c.186-19G>A	NA	NA	NA	9.11	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089944	G	A	NA	NO	synonymous	ENST00000276616.4:c.225G>A	NA	L75L	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	8.402	NA	NA	NA	NA	NA	0.00000	
ATM	11	108089970	C	T	rs587781937	NO	missense	ENST00000276616.4:c.251C>T	ENSP00000276616.4:p.Ala84Val	A84V	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	23.2	benign	0.119	deleterious	0.05	0	0.00000	
ATM	11	108089994	G	C	NA	NO	missense	ENST00000276616.4:c.275A>C	ENSP00000276616.4:p.Gln92Pro	Q92P	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	25.5	probably_damaging	0.953	deleterious	0.01	0	0.00000	
ATM	11	108089993	A	C	rs756969590	NO	missense	ENST00000276616.4:c.274A>C	ENSP00000276616.4:p.Lys92Gln	K92Q	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	25.6	probably_damaging	0.998	deleterious	0.02	0	0.00000	
ATM	11	108100010	C	T	rs774676210	NO	synonymous	ENST00000276616.4:c.291C>T	NA	P97I	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	13.08	NA	NA	NA	NA	NA	0.00006	
ATM	11	108100014	A	G	rs137842685,CM991047	NO	missense	ENST00000276616.4:c.295A>G	ENSP00000276616.4:p.Ser99Gly	S99G	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	23.3	benign	0.168	deleterious	0.03	0	0.00041	
ATM	11	108100019	G	A	CM010777,rs142535238	NO	missense	ENST00000276616.4:c.320G>A	ENSP00000276616.4:p.Cys107Tyr	C107Y	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	16.18	benign	0.158	tolerated	0.46	0	0.00000	
ATM	11	108106371	T	C	NA	NO	intron	ENST00000276616.4:c.332-2815T>C	NA	NA	NA	1.662	NA	NA	NA	NA	NA	0.00000	
ATM	11	108106379	T	A	NA	NO	intron	ENST00000276616.4:c.332-1817>A	NA	NA	NA	10.7	NA	NA	NA	NA	NA	0.00000	
ATM	11	108106399	G	A	rs146382972	NO	missense	ENST00000276616.4:c.334G>A	ENSP00000276616.4:p.Ala112Thr	A112T	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	23	benign	0.41	tolerated	0.06	0	0.00000	
ATM	11	108106402	T	C	rs147309948,CM989940	NO	synonymous	ENST00000276616.4:c.336C>T	ENSP00000276616.4:p.Pro113Leu	P113L	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	33	probably_damaging	1	deleterious	0	65	0.00000	
ATM	11	108106435	A	G	CD000908,rs145590873	NO	missense	ENST00000276616.4:c.370A>G	ENSP00000276616.4:p.Ile124Val	I124V	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	0.491	benign	0.006	tolerated	1	0	0.00024	
ATM	11	108106437	C	A	rs774459195	NO	synonymous	ENST00000276616.4:c.372C>A	NA	I124I	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	17.97	NA	NA	NA	NA	NA	0.00006	
ATM	11	108106443	T	A	rs587781449,rs2234997,CM077897,COSM2249	NO	missense	ENST00000276616.4:c.378T>A	ENSP00000276616.4:p.Asp126Glu	D126E	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	4.635	benign	0.006	tolerated	1	0	0.00024	
ATM	11	108106463	G	A	rs728881330	NO	missense	ENST00000276616.4:c.398A>G	ENSP00000276616.4:p.Asn133Ser	N133S	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	0.001	benign	0.003	tolerated	1	0	0.00024	
ATM	11	108106466	G	A	rs771166271	NO	missense	ENST00000276616.4:c.401G>A	ENSP00000276616.4:p.Gly134Asp	G134D	Plant_domain:PF13640,PTIR11139.PTIR11139:SF166	14.5	possibly_damaging	0.493	tolerated	0.42	0	0.00000	
ATM	11	108106565	T	C	rs587781375	NO	splice_intron	ENST00000276616.4:c.496+47:C	NA	NA	NA	4.514	NA	NA	NA	NA	NA	0.03115	
ATM	11	108114609	G	A	NA	NO	intron	ENST00000276616.4:c.497-712:A	NA	NA	NA	2.263	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114632	T	G	rs2235001	NO	intron	ENST00000276616.4:c.497-687:G	NA	NA	NA	0.205	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114637	A	G	NA	NO	intron	ENST00000276616.4:c.497-683:G	NA	NA	NA	1.119	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114655	C	G	NA	NO	intron	ENST00000276616.4:c.497-25:C>G	NA	NA	NA	1.385	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114666	T	A	rs757147178	NO	intron	ENST00000276616.4:c.497-147>A	NA	NA	NA	2.356	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114680	A	T	NA	NO	missense_splice	ENST00000276616.4:c.497A>T	ENSP00000276616.4:p.Glu166Val	E166V	PTIR11139.PTIR11139:SF166	23.2	benign	0.219	deleterious	0.01	15	0.00000	
ATM	11	108114727	G	C	CM051538,rs12187307	NO	missense	ENST00000276616.4:c.502G>C	ENSP00000276616.4:p.Val168Leu	V182L	PTIR11139.PTIR11139:SF166	12.39	benign	0.009	tolerated	0.2	0	0.00023	
ATM	11	108114733	G	A	CM093706,rs753806542	NO	missense	ENST00000276616.4:c.509G>A	ENSP00000276616.4:p.Gly197Glu	G197E	PTIR11139.PTIR11139:SF166	27.9	probably_damaging	0.999	deleterious	0.01	65	0.00000	
ATM	11	108114742	G	T	rs147309948,CM989940	NO	synonymous	ENST00000276616.4:c.518C>T	NA	D200D	PTIR11139.PTIR11139:SF166	14.94	NA	NA	NA	NA	NA	0.00438	
ATM	11	108114793	G	A	rs147915571,CM054524,CSM401768,COSMN0	NO	missense	ENST00000276616.4:c.610G>A	ENSP00000276616.4:p.Gly204Arg	G204R	PTIR11139.PTIR11139:SF166	23.7	possibly_damaging	0.609	tolerated	0.44	0	0.00027	
ATM	11	108114829	G	A	rs2235002	NO	missense	ENST00000276616.4:c.646G>T	ENSP00000276616.4:p.Ala216Ser	A216S	PTIR11139.PTIR11139:SF166	20.8	benign	0.42	tolerated	0.23	0	0.00003	
ATM	11	108114840	T	C	rs2235003	NO	synonymous	ENST00000276616.4:c.657T:C	NA	C219C	PTIR11139.PTIR11139:SF166	0.007	NA	NA	NA	NA	NA	0.00027	
ATM	11	108114859	T	A	NA	NO	intron	ENST00000276616.4:c.662+147>A	NA	NA	NA	4.518	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114883	T	C	rs2235004	NO	intron	ENST00000276616.4:c.662-183:C>	NA	NA	NA	5.368	NA	NA	NA	NA	NA	0.00000	
ATM	11	108114887	T	A	rs762680194	NO	intron	ENST00000276616.4:c.662+427>A	NA	NA	NA	7.911	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115483	A	G	NA	NO	synonymous	ENST00000276616.4:c.663-23A>G	NA	NA	NA	1.573	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115522	A	G	rs149853092,CM000650	NO	missense	ENST00000276616.4:c.676A>G	ENSP00000276616.4:p.Lys226Glu	K226E	PTIR11139.PTIR11139:SF166	23.5	benign	0.157	deleterious	0.02	0	0.00021	
ATM	11	108115580	T	C	CD034117660	NO	missense	ENST00000276616.4:c.725T:C	ENSP00000276616.4:p.Leu243Ser	L243S	PTIR11139.PTIR11139:SF166	15.13	benign	0.006	tolerated	0.21	0	0.00000	
ATM	11	108115587	C	T	rs2218674	NO	synonymous	ENST00000276616.4:c.732C>T	NA	V245V	PTIR11139.PTIR11139:SF166	11.67	NA	NA	NA	NA	NA	0.03115	
ATM	11	108115600	C	T	rs728821016,CM001038,CS991299,COSM1732YES		stop_gained	ENST00000276616.4:c.746C>T	ENSP00000276616.4:p.Arg250Ter	R250*	PTIR11139.PTIR11139:SF166	36	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115601	G	A	CM092564,rs61125940	NO	missense	ENST00000276616.4:c.747G>A	ENSP00000276616.4:p.Arg250Gln	R250Q	PTIR11139.PTIR11139:SF166	13.4	benign	0.02	tolerated	0.36	0	0.00015	
ATM	11	108115639	CT	C	rs774609577	YES	frameshift_feature_truncation	ENST00000276616.4:c.790delT	ENSP00000276616.4:p.Tyr264ReferTer12	L263X	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	31	NA	NA	NA	NA	NA	0.00003	
ATM	11	108115657	CAT	C	NA	YES	frameshift_feature_truncation	ENST00000276616.4:c.807_808delTA	ENSP00000276616.4:p.His269GlnfsTer3	H269X	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	34	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115663	C	A	rs730881339	NO	missense	ENST00000276616.4:c.811C>A	ENSP00000276616.4:p.Leu271Ile	L271I	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	18.06	benign	0.32	tolerated	0.24	0	0.00003	
ATM	11	108115673	CT	C	NA	YES	frameshift_feature_truncation	ENST00000276616.4:c.824delT	ENSP00000276616.4:p.Leu275Ter	S276X	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	23.9	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115698	A	T	rs754008850	NO	missense	ENST00000276616.4:c.846A>T	ENSP00000276616.4:p.Leu282Phe	L282F	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	6.125	benign	0.009	tolerated	1	0	0.00000	
ATM	11	108115703	A	T	NA	NO	intron	ENST00000276616.4:c.851A>T	ENSP00000276616.4:p.Glu284Leu	Q284L	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	16.06	benign	0.003	tolerated	0.29	0	0.00000	
ATM	11	108115706	T	G	NA	NO	missense	ENST00000276616.4:c.854T>G	ENSP00000276616.4:p.Leu285Arg	L285R	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	28.5	probably_damaging	0.977	deleterious	0	65	0.00000	
ATM	11	108115718	C	A	rs15261362	NO	missense	ENST00000276616.4:c.866T>C	ENSP00000276616.4:p.Ile289Thr	I289T	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	22.8	benign	0.097	deleterious	0.03	0	0.00000	
ATM	11	108115728	G	A	rs755860432	NO	synonymous	ENST00000276616.4:c.876G>A	NA	P292P	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	12.23	NA	NA	NA	NA	NA	0.00003	
ATM	11	108115738	T	G	rs1800735	NO	intron	ENST00000276616.4:c.901-2T:G	NA	NA	NA	5.574	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115841	T	C	NA	NO	intron	ENST00000276616.4:c.902-1017:C	NA	NA	NA	3.225	NA	NA	NA	NA	NA	0.00000	
ATM	11	108115691	G	A	rs202088661,CM991482	NO	missense_splice	ENST00000276616.4:c.902G>A	ENSP00000276616.4:p.Gly301Asp	G301D	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	23.8	benign	0.374	deleterious	0.01	65	0.00015	
ATM	11	108117714	A	T	NA	YES	stop_gained	ENST00000276616.4:c.924A>T	ENSP00000276616.4:p.Tyr308*	R308*	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	36	NA	NA	NA	NA	NA	0.00192	
ATM	11	108117735	T	C	rs142317485	NO	missense	ENST00000276616.4:c.946T>C	ENSP00000276616.4:p.Tyr316His	Y316H	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	27.3	probably_damaging	0.986	deleterious	0	35	0.00009	
ATM	11	108117747	G	A	NA	NO	missense	ENST00000276616.4:c.958G>A	ENSP00000276616.4:p.Val320Met	V320M	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	24.9	possibly_damaging	0.513	deleterious	0.01	0	0.00000	
ATM	11	108117767	A	G	NA	NO	missense	ENST00000276616.4:c.978A>G	ENSP00000276616.4:p.Ile326Met	I326M	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	23.2	possibly_damaging	0.954	tolerated	0.05	0	0.00000	
ATM	11	108117787	C	T	rs28904919	NO	missense	ENST00000276616.4:c.998C>T	ENSP00000276616.4:p.Ser333Phe	S333F	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	25.9	possibly_damaging	0.602	deleterious	0.01	15	0.00200	
ATM	11	108117798	C	T	CM091043,rs138398778,COSM2123,COSM2NO		missense	ENST00000276616.4:c.1009C>T	ENSP00000276616.4:p.Arg337Cys	R337C	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	35	probably_damaging	0.999	deleterious	0.5	0	0.00027	
ATM	11	108117799	G	A	rs202104835,COSM1235400,COSM2301,COSMN	NO	missense	ENST00000276616.4:c.1010G>A	ENSP00000276616.4:p.Arg337His	R337H	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	34	probably_damaging	0.997	deleterious	0.02	0	0.00027	
ATM	11	108117840	G	A	CSM092783,COSM1583367	NO	missense	ENST00000276616.4:c.1051G>A	ENSP00000276616.4:p.Asp351Asn	D351N	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	27	probably_damaging	0.983	tolerated	0.05	15	0.00000	
ATM	11	108117852	G	A	NA	NO	intron	ENST00000276616.4:c.1065+98G>A	NA	NA	NA	4.007	NA	NA	NA	NA	NA	0.03115	
ATM	11	108119581	C	T	NA	NO	intron	ENST00000276616.4:c.1066-796:C>	ENSP00000276616.4:p.Ala356Val	A356V	PTIR11139.PTIR11139:SF166	2.072	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119586	A	G	NA	NO	intron	ENST00000276616.4:c.1066-764:A>G	NA	NA	NA	1.944	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119595	A	G	NA	NO	intron	ENST00000276616.4:c.1066-654A>G	NA	NA	NA	1.083	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119599	C	T	NA	NO	intron	ENST00000276616.4:c.1066-614C> T	NA	NA	NA	0.777	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119602	C	T	NA	NO	intron	ENST00000276616.4:c.1066-586C>T	NA	NA	NA	0.695	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119603	G	A	rs4987935	NO	intron	ENST00000276616.4:c.1066-576G>A	NA	NA	NA	0.647	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119654	T	G	rs201618625,CS000846,COSM758349	NO	splice_intron	ENST00000276616.4:c.1066-67:G>	NA	NA	NA	11.93	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119667	A	G	rs149636614	NO	missense	ENST00000276616.4:c.1073A>G	ENSP00000276616.4:p.Asn358Ser	N358S	Superfamily_domains:SSF48371,PTIR11139.PTIR11139:SF166	0.284	benign	0.001	tolerated	0.95	0	0.00006	
ATM	11	108119687	G	T	NA	YES	stop_gained	ENST00000276616.4:c.1093G>T	ENSP00000276616.4:p.Glu365Ter	E365*	PTIR11139.PTIR11139:SF166	38	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119752	A	TA	NA	YES	frameshift_feature_elongation	ENST00000276616.4:c.1109dupA	ENSP00000276616.4:p.Tyr370Ter	Y370*	PTIR11139.PTIR11139:SF166	25.7	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119704	C	T	rs776178608,CM013634,CD991597	NO	synonymous	ENST00000276616.4:c.1110C>T	NA	Y370Y	PTIR11139.PTIR11139:SF166	0.158	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119732	T	A	rs148820885	NO	missense	ENST00000276616.4:c.1138T>A	ENSP00000276616.4:p.Tyr380Asn	Y380N	PTIR11139.PTIR11139:SF166	0.213	benign	0.021	tolerated	0.38	0	0.00003	
ATM	11	108119740	C	T	NA	NO	synonymous	ENST00000276616.4:c.1146C>T	NA	Y382Y	PTIR11139.PTIR11139:SF166	8.232	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119779	G	A	rs1800727	NO	synonymous	ENST00000276616.4:c.1176C>A	NA	G392G	PTIR11139.PTIR11139:SF166	12.47	NA	NA	NA	NA	NA	0.00033	
ATM	11	108119791	C	T	NA	NO	synonymous	ENST00000276616.4:c.1197C>T	NA	H399H	PTIR11139.PTIR11139:SF166	13	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119823	C	C	CM051782,rs61287836,COSM1350756,COSM2NO		missense	ENST00000276616.4:c.1229T>C	ENSP00000276616.4:p.Val410Ala	V410A	PTIR11139.PTIR11139:SF166	23.2	benign	0.22	deleterious	0.01	25	0.00649	
ATM	11	108119839	T	TA	rs771187419	NO	intron_feature_elongation	ENST00000276616.4:c.1235+11dupA	NA	NA	NA	10.84	NA	NA	NA	NA	NA	0.00000	
ATM	11	108119949	T	G	rs782656752	NO	missense	ENST00000276616.4:c.1235+285T>G	NA	NA	NA	7.632	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121464	A	G	rs4987943	NO	synonymous	ENST00000276616.4:c.2254A>G	ENSP00000276616.4:p.Ser752Gly	S752G	PTIR11139.PTIR11139:SF166	0.816	NA	NA	NA	NA	NA	0.03115	
ATM	11	108121464	T	C	rs35578748	NO	synonymous	ENST00000276616.4:c.1272T:C	NA	P424P	PTIR11139.PTIR11139:SF166	3.835	NA	NA	NA	NA	NA	0.00091	
ATM	11	108121479	CTG	CA	NA	YES	frameshift_feature_truncation	ENST00000276616.4:c.1288_1291delTG	ENSP00000276616.4:p.Cys430Ter	C430X	PTIR11139.PTIR11139:SF166	24.8	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121524	C	A	rs763361384	NO	synonymous	ENST00000276616.4:c.1332C>A	NA	P444P	Low_complexity_(Seg):sp.PTIR11139.PTIR11139:SF166	13.76	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121527	A	G	NA	NO	synonymous	ENST00000276616.4:c.1335A>G	NA	Q445Q	Low_complexity_(Seg):sp.PTIR11139.PTIR11139:SF166	0.486	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121531	C	T	rs587798815,CM960898,COSM3733248,COSNYES		stop_gained	ENST00000276616.4:c.1339C>T	ENSP00000276616.4:p.Arg447Ter	R447*	PTIR11139.PTIR11139:SF166	39	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121543	T	C	rs28171992?,COSM1321925,COSM1321924	NO	missense	ENST00000276616.4:c.1351T>C	ENSP00000276616.4:p.Ser451Pro	S451C	PTIR11139.PTIR11139:SF166	33	probably_damaging	0.997	deleterious	0.02	0	0.00000	
ATM	11	108121546	AC	A	rs201333862,rs587781776,CD982449	YES	frameshift_feature_truncation	ENST00000276616.4:c.1355delAC	ENSP00000276616.4:p.Thr452AsnfsTer21	T452X	PTIR11139.PTIR11139:SF166	24.8	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121546	A	G	NA	NO	missense	ENST00000276616.4:c.1354A>G	ENSP00000276616.4:p.Thr452Ala	T452A	PTIR11139.PTIR11139:SF166	4.062	benign	0.005	tolerated	0.41	0	0.00006	
ATM	11	108121551	A	T	NA	NO	synonymous	ENST00000276616.4:c.1359A>T	NA	P453P	PTIR11139.PTIR11139:SF166	0.007	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121559	T	TA	NA	YES	frameshift_feature_elongation	ENST00000276616.4:c.1366dupA	ENSP00000276616.4:p.Arg457TfsTer30	L456LX	PTIR11139.PTIR11139:SF166	29.2	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121570	A	C	rs587782729	NO	missense	ENST00000276616.4:c.1378A>C	ENSP00000276616.4:p.Thr460Pro	T460P	PTIR11139.PTIR11139:SF166	8.091	benign	0.078	tolerated	0.22	0	0.00006	
ATM	11	108121591	CAA	C	rs587781147,COSM758338,COSM1289451	YES	frameshift_feature_truncation	ENST00000276616.4:c.1402-1403delAA	ENSP00000276616.4:p.Lys468ArgfsTer18	K468X	PTIR11139.PTIR11139:SF166	28.3	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121608	A	G	CSM012737	NO	synonymous	ENST00000276616.4:c.1416A>G	NA	L472L	PTIR11139.PTIR11139:SF166	2.68	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121612	A	C	rs150240037	NO	missense	ENST00000276616.4:c.1420A>C	ENSP00000276616.4:p.Lys474Pro	L474P	PTIR11139.PTIR11139:SF166	23.8	probably_damaging	0.996	deleterious	0.02	0	0.00003	
ATM	11	108121635	A	C	rs753800755	NO	missense	ENST00000276616.4:c.1443A>C	ENSP00000276616.4:p.Lys481Phe	L481F	PTIR11139.PTIR11139:SF166	8.643	benign	0.058	tolerated	0.71	0	0.00000	
ATM	11	108121636	A	G	rs202173660,COSM922707,COSM1585363	NO	missense	ENST00000276616.4:c.1444A>G	ENSP00000276616.4:p.Lys482Glu	K482Q	PTIR11139.PTIR11139:SF166	17.59	benign	0.242	tolerated	0.08	0	0.00024	
ATM	11	108121656	G	T	rs775597049	NO	missense	ENST00000276616.4:c.1464G>T	ENSP00000276616.4:p.Trp488Cys	W488C	PTIR11139.PTIR11139:SF166	23.7	benign	0.242	deleterious	0.05	65	0.00009	
ATM	11	108121687	C	T	CS082617912	YES	stop_gained	ENST00000276616.4:c.1495C>T	ENSP00000276616.4:p.Gln499Ter	Q499*	PTIR11139.PTIR11139:SF166	38	NA	NA	NA	NA	NA	0.00000	
ATM	11	108121708	G	T	rs587778816	NO	missense	ENST00000276616.4:c.1516G>T	ENSP00000276616.4:p.Gly506Cys	G506C	PTIR11139.PTIR11139:SF166	29	possibly_damaging	0.67	deleterious	0.01	15	0.00021	
ATM	11	108121718	G	A	NA	NO	missense	ENST00000276616.4:c.1526G>A	ENSP00000276616.4:p.Gly509Glu	G509E	PTIR11139.PTIR11139:SF166	16.19	benign	0.027	tolerated	0.15	0	0.00006	
ATM	11	108121733	G	A	CM024583,rs2235000	NO	missense	ENST00000276616.4:c.1541G>

ATM	11	108139307	T	C	rs756618769	NO	intron	ENST00000227616:4:c.2639-30T=C	NA	NA	NA	6.648	NA	NA	NA	NA	NA	0.00000
ATM	11	108139120	G	T	rs2234994	NO	intron	ENST00000227616:4:c.2639-17G=T	NA	NA	NA	6.735	NA	NA	NA	NA	NA	0.00000
ATM	11	108139177	A	G	rs139316519	NO	synonymous	ENST00000227616:4:c.2670A=G	NA	Q893Q	PTIR1139;PTIR1139:SF86	2.565	NA	NA	NA	NA	NA	0.00093
ATM	11	108139183	A	G	rs2218687	NO	synonymous	ENST00000227616:4:c.2685A=G	L895L	L895L	Low_complexity_(Seg):seg,PTIR1139;PTIR1139:SF86	1.417	NA	NA	NA	NA	NA	0.00024
ATM	11	108139187	T	A	rs147122522;CD000911	NO	missense	ENST00000227616:4:c.2689T=A	ENSP00000227616:4:p.Phe897Ile	F897I	Low_complexity_(Seg):seg,PTIR1139;PTIR1139:SF86	9.477	benign	0.001	tolerated	0.25	0	0.00000
ATM	11	108139221	T	C	rs754738085	NO	missense	ENST00000227616:4:c.2723T=C	ENSP00000227616:4:p.Val908Ala	V908A	PTIR1139;PTIR1139:SF86	8.54	benign	0.003	tolerated	0.85	0	0.00000
ATM	11	108139236	C	A	NA	NO	missense	ENST00000227616:4:c.2738C=A	ENSP00000227616:4:p.Thr913Asn	T913N	PTIR1139;PTIR1139:SF86	0.086	benign	0.001	tolerated	0.5	0	0.00000
ATM	11	108139302	C	T	rs2218788	NO	missense	ENST00000227616:4:c.2804C=T	ENSP00000227616:4:p.Pro935Ser	P935S	PTIR1139;PTIR1139:SF86	8.915	benign	0.003	tolerated	0.3	0	0.00027
ATM	11	108139303	G	C	rs1954812;COSM592	NO	synonymous	ENST00000227616:4:c.2805G=C	NA	T935T	PTIR1139;PTIR1139:SF86	0.701	NA	NA	NA	NA	NA	0.00093
ATM	11	108139306	A	G	rs147791214	NO	synonymous	ENST00000227616:4:c.2808A=G	NA	L936L	PTIR1139;PTIR1139:SF86	0.06	NA	NA	NA	NA	NA	0.00000
ATM	11	108139322	C	T	rs121868;COSM148656	NO	missense	ENST00000227616:4:c.2824C=T	ENSP00000227616:4:p.Leu942Phe	L942F	PTIR1139;PTIR1139:SF86	23.2	possibly_damaging	0.847	deleterious	0.02	0	0.00000
ATM	11	108139345	C	G	rs576168823;COSM237819	NO	intron	ENST00000227616:4:c.2838+9C=G	NA	NA	NA	3.099	NA	NA	NA	NA	NA	0.00000
ATM	11	108139346	G	A	NA	NO	intron	ENST00000227616:4:c.2838+10G=A	NA	NA	NA	0.566	NA	NA	NA	NA	NA	0.00000
ATM	11	108139364	T	TA	rs2234995	NO	intron	ENST00000227616:4:c.2838+28_2838+29insA	NA	NA	NA	3.857	NA	NA	NA	NA	NA	0.00000
ATM	11	108139376	G	C	rs745417415	NO	intron	ENST00000227616:4:c.2838+40G=C	NA	NA	NA	3.873	NA	NA	NA	NA	NA	0.00000
ATM	11	108139382	G	T	rs574141554	NO	intron	ENST00000227616:4:c.2838+46G=T	NA	NA	NA	6.19	NA	NA	NA	NA	NA	0.00000
ATM	11	108139396	A	G	rs748534458	NO	intron	ENST00000227616:4:c.2838+60A=G	NA	NA	NA	9.165	NA	NA	NA	NA	NA	0.00000
ATM	11	108139397	T	A	rs4454103	NO	intron	ENST00000227616:4:c.2838+61T=A	NA	NA	NA	10.64	NA	NA	NA	NA	NA	0.00000
ATM	11	108139405	C	CA	NA	NO	intron	ENST00000227616:4:c.2838-7dupA	NA	NA	NA	5.538	NA	NA	NA	NA	NA	0.00000
ATM	11	108139471	C	T	rs761953913	NO	intron	ENST00000227616:4:c.2839-16C=T	NA	NA	NA	3.354	NA	NA	NA	NA	NA	0.00000
ATM	11	108141816	C	A	COSM1492485;COSM1492404	NO	missense	ENST00000227616:4:c.2864C=A	ENSP00000227616:4:p.Pro955His	P955H	PTIR1139;PTIR1139:SF86	29.4	probably_damaging	0.986	deleterious	0.01	0	0.00000
ATM	11	108141841	G	A	rs35988825	NO	missense	ENST00000227616:4:c.2890G=A	ENSP00000227616:4:p.Met963Ile	M963I	PTIR1139;PTIR1139:SF86	16.68	benign	0.000	tolerated	0.18	0	0.00000
ATM	11	108141871	A	G	rs587778829	NO	splice_synonymous	ENST00000227616:4:c.2919A=G	NA	L973	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	12.03	NA	NA	NA	NA	NA	0.00000
ATM	11	108141889	C	A	rs558376224	NO	intron	ENST00000227616:4:c.2921+16C=A	NA	NA	NA	12.84	NA	NA	NA	NA	NA	0.00000
ATM	11	108141891	T	TA	rs575967175	NO	intron_feature_elongation	ENST00000227616:4:c.2921-19dupA	NA	NA	NA	10.58	NA	NA	NA	NA	NA	0.00000
ATM	11	108141894	G	GT	rs755113481	NO	intron_feature_elongation	ENST00000227616:4:c.2921-23dupT	NA	NA	NA	7.419	NA	NA	NA	NA	NA	0.00000
ATM	11	108141897	A	G	rs763051555	NO	intron	ENST00000227616:4:c.2921+24A=G	NA	NA	NA	4.58	NA	NA	NA	NA	NA	0.00000
ATM	11	108141899	G	A	NA	NO	intron	ENST00000227616:4:c.2921-26G=A	NA	NA	NA	0.262	NA	NA	NA	NA	NA	0.00000
ATM	11	108141913	T	A	NA	NO	intron	ENST00000227616:4:c.2921+40T=A	NA	NA	NA	4.231	NA	NA	NA	NA	NA	0.00000
ATM	11	108141924	TG	T	NA	NO	intron_feature_truncation	ENST00000227616:4:c.2921+52delG	NA	NA	NA	9.711	NA	NA	NA	NA	NA	0.00000
ATM	11	108141929	T	G	NA	NO	intron	ENST00000227616:4:c.2922-49T=G	NA	NA	NA	1.953	NA	NA	NA	NA	NA	0.00000
ATM	11	108141937	T	A	rs748534548	NO	intron	ENST00000227616:4:c.2922-61T=A	NA	NA	NA	3.092	NA	NA	NA	NA	NA	0.00000
ATM	11	108141941	C	A	NA	NO	intron	ENST00000227616:4:c.2922-65C=A	NA	NA	NA	4.429	NA	NA	NA	NA	NA	0.00000
ATM	11	108141943	T	G	rs759326892	NO	intron	ENST00000227616:4:c.2922-357T=G	NA	NA	NA	1.669	NA	NA	NA	NA	NA	0.00000
ATM	11	108141980	A	G	rs730881154;COSM910495	NO	missense_splice	ENSP00000227616:4:p.Asn975Ser	ENSP00000227616:4:p.Asn975Ser	N975S	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	8.852	benign	0.001	tolerated	0.42	0	0.00000
ATM	11	108141988	T	C	rs139552233;COSM922715;COSM22828;COSN	NO	missense	ENST00000227616:4:c.2912T=C	ENSP00000227616:4:p.Ser970Pro	S970P	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	26.8	possibly_damaging	0.797	deleterious	0.01	65	0.00147
ATM	11	108141989	T	G	rs568678134;COSM1150813;COSM1350812	NO	synonymous	ENST00000227616:4:c.2913T=G	NA	S970S	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	1.101	NA	NA	NA	NA	NA	0.00015
ATM	11	108141995	A	T	NA	NO	missense	ENST00000227616:4:c.2919A=T	ENSP00000227616:4:p.Tyr980Phe	Y980F	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	24	possibly_damaging	0.501	tolerated	0.08	0	0.00000
ATM	11	108141998	G	A	rs755898387	NO	missense	ENST00000227616:4:c.2922G=A	ENSP00000227616:4:p.Arg981His	R981H	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	23.2	benign	0.017	tolerated	0.07	25	0.00000
ATM	11	108142000	C	T	rs587778830;COSM1297603;COSM1297602	NO	missense	ENST00000227616:4:c.2944C=T	ENSP00000227616:4:p.Arg982Cys	R982C	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	34	possibly_damaging	0.893	deleterious	0	35	0.00006
ATM	11	108142004	A	T	NA	NO	missense	ENST00000227616:4:c.2948A=T	ENSP00000227616:4:p.Asp983Val	D983V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	23.4	benign	0.273	deleterious	0.01	0	0.00000
ATM	11	108142070	A	G	rs146531614	NO	missense	ENST00000227616:4:c.3014A=G	ENSP00000227616:4:p.Asn1005Ser	N1005S	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.001	benign	0.001	tolerated	0.97	0	0.00078
ATM	11	108142072	A	G	rs148093195	NO	missense	ENST00000227616:4:c.3016A=G	ENSP00000227616:4:p.Met1006Val	M1006V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.001	benign	0.001	tolerated	0.62	0	0.00018
ATM	11	108142074	G	T	NA	NO	missense	ENST00000227616:4:c.3018G=T	ENSP00000227616:4:p.Met1006Ile	M1006I	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.105	benign	0.004	tolerated	0.46	0	0.00075
ATM	11	108142307	G	T	NA	NO	missense	ENST00000227616:4:c.3031G=T	ENSP00000227616:4:p.Gln1011His	Q1011H	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	11.5	benign	0.008	tolerated	0.33	0	0.00000
ATM	11	108142341	C	G	NA	NO	splice_intron	ENST00000227616:4:3077+8C=G	NA	NA	NA	4.917	NA	NA	NA	NA	NA	0.00000
ATM	11	108142358	C	T	rs773984258	NO	intron	ENST00000227616:4:3077+28C=T	NA	NA	NA	3.553	NA	NA	NA	NA	NA	0.00000
ATM	11	108142360	A	G	rs200922643	NO	intron	ENST00000227616:4:3077+27A=G	NA	NA	NA	3.704	NA	NA	NA	NA	NA	0.00000
ATM	11	108142382	C	T	rs464677	NO	intron	ENST00000227616:4:3078-7T=C	NA	NA	NA	4.058	NA	NA	NA	NA	NA	0.00000
ATM	11	108142384	A	G	NA	NO	intron	ENST00000227616:4:3078-75A=G	NA	NA	NA	4.808	NA	NA	NA	NA	NA	0.00000
ATM	11	108142427	T	C	NA	NO	intron	ENST00000227616:4:3078-227T=C	NA	NA	NA	1.06	NA	NA	NA	NA	NA	0.00000
ATM	11	108142558	G	A	rs750663117;CS992442	YES	splice_acceptor	ENST00000227616:4:3079-1G=A	NA	NA	NA	26.4	NA	NA	NA	NA	NA	0.00003
ATM	11	108142561	A	G	NA	NO	missense_splice	ENST00000227616:4:3080A=G	ENSP00000227616:4:p.His1027Arg	H1027R	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	12.5	benign	0.003	tolerated	0.35	0	0.00000
ATM	11	108142363	C	A	NA	NO	missense	ENST00000227616:4:3082C=A	ENSP00000227616:4:p.Leu1028Ile	L1028I	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	23.7	possibly_damaging	0.566	tolerated	0.05	0	0.00000
ATM	11	108142366	AC	A	NA	YES	frameshift_feature_truncation	ENST00000227616:4:3086delC	ENSP00000227616:4:p.Thr1029Lysfs*Ter10	T1029X	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	35	NA	NA	NA	NA	NA	0.00000
ATM	11	108142393	G	A	NA	NO	missense	ENST00000227616:4:3112G=A	ENSP00000227616:4:p.Val1038Ile	V1038I	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	26.9	probably_damaging	0.94	deleterious	0	25	0.00000
ATM	11	108142395	A	T	rs374451781	NO	synonymous	ENST00000227616:4:3114A=T	ENSP00000227616:4:p.Val1039Val	V1039V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	1.385	NA	NA	NA	NA	NA	0.00000
ATM	11	108142399	A	G	rs200287;COSM32486	NO	missense	ENST00000227616:4:3116A=G	ENSP00000227616:4:p.Met1040Val	M1040V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.019	benign	0.001	tolerated	1	0	0.00000
ATM	11	108142313	T	C	NA	NO	synonymous	ENST00000227616:4:3112T=C	NA	N1044N	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.158	NA	NA	NA	NA	NA	0.00000
ATM	11	108142331	T	C	rs1002859	NO	synonymous	ENST00000227616:4:3135T=C	NA	L1050L	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	4.799	NA	NA	NA	NA	NA	0.00105
ATM	11	108142367	A	G	NA	NO	intron	ENST00000227616:4:3153-13A=G	NA	NA	NA	4.394	NA	NA	NA	NA	NA	0.00000
ATM	11	108142382	T	A	NA	NO	intron	ENST00000227616:4:3154-57T=A	NA	NA	NA	11.69	NA	NA	NA	NA	NA	0.00000
ATM	11	108142422	T	C	rs758643104	NO	intron	ENST00000227616:4:3154-237T=C	NA	NA	NA	5.864	NA	NA	NA	NA	NA	0.00000
ATM	11	108142437	C	A	rs781387283	NO	intron	ENST00000227616:4:3154-12C=A	NA	NA	NA	8.895	NA	NA	NA	NA	NA	0.00000
ATM	11	108142444	C	T	rs575197759	NO	splice_intron	ENST00000227616:4:3154-5C=T	NA	NA	NA	8.171	NA	NA	NA	NA	NA	0.00000
ATM	11	108142445	G	A	rs199543113	NO	splice_intron	ENST00000227616:4:3154-6G=A	NA	NA	NA	5.041	NA	NA	NA	NA	NA	0.00000
ATM	11	108142450	C	T	NA	NO	missense_splice	ENST00000227616:4:3155C=T	ENSP00000227616:4:p.Ala1052Val	A1052V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	4.126	benign	0.007	tolerated	1	0	0.00000
ATM	11	108142456	G	A	rs587783365;rs1800057;COSM21827	NO	missense	ENST00000227616:4:3161G=A	ENSP00000227616:4:p.Pro1054Leu	P1054R	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	26.5	probably_damaging	0.982	deleterious	0	65	0.01159
ATM	11	108142470	G	A	rs370828631	NO	missense	ENST00000227616:4:3175G=A	ENSP00000227616:4:p.Ala1059Thr	A1059T	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	16.44	benign	0.082	tolerated	0.28	0	0.00000
ATM	11	108142486	T	A	NA	NO	missense	ENST00000227616:4:3191T=C	ENSP00000227616:4:p.Met1064Ala	M1064T	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.044	benign	0.028	tolerated	0.16	0	0.00000
ATM	11	108142514	A	G	CD052443;rs752849892	YES	splice_acceptor	ENST00000227616:4:3219-1G=A	NA	V1073V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	10.47	NA	NA	NA	NA	NA	0.00003
ATM	11	108142517	A	G	rs144111672;COSM1726571;COSM1726570	NO	missense	ENST00000227616:4:3222A=G	ENSP00000227616:4:p.Ser1074Gly	S1074G	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	0.057	benign	0.001	tolerated	1	0	0.00000
ATM	11	108142540	CATC	CTGAT	NA	YES	frameshift_feature_elongation	ENST00000227616:4:3240_3244delTCAtcaTGAT	ENSP00000227616:4:p.His1083AspfsTer13	QS1082-10R3HDX	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	35	NA	NA	NA	NA	NA	0.00000
ATM	11	108142560	G	T	rs730881158	NO	missense	ENST00000227616:4:3265G=T	ENSP00000227616:4:p.Ala1089Ser	A1089S	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	18	benign	0.035	tolerated	0.21	0	0.00000
ATM	11	108142601	A	C	rs763039403	NO	intron	ENST00000227616:4:3284-22A=C	NA	NA	NA	4.479	NA	NA	NA	NA	NA	0.00000
ATM	11	108142608	T	A	rs778294782	NO	intron	ENST00000227616:4:3285-30T=A	NA	NA	NA	6.498	NA	NA	NA	NA	NA	0.00000
ATM	11	108142698	G	A	NA	NO	intron	ENST00000227616:4:3285-206G=A	NA	NA	NA	0.153	NA	NA	NA	NA	NA	0.00000
ATM	11	108142807	CT	TC	rs1798757	NO	intron_feature_truncation	ENST00000227616:4:3285-9delT	NA	NA	NA	7.431	NA	NA	NA	NA	NA	0.00000
ATM	11	108142825	C	A	rs773964990	NO	missense	ENST00000227616:4:3292C=A	ENSP00000227616:4:p.Gln1098Lys	Q1098K	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	10.53	benign	0.028	tolerated	1	0	0.00000
ATM	11	108142828	G	T	rs372960951;CM009149?	NO	missense	ENST00000227616:4:3295G=T	ENSP00000227616:4:p.Arg1099Ser	D1099Y	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	21	benign	0.011	deleterious	0.01	0	0.00000
ATM	11	108142833	G	A	rs587780621	NO	synonymous	ENST00000227616:4:3300G=A	ENSP00000227616:4:p.Ala1100Ala	T1100T	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	6.657	NA	NA	NA	NA	NA	0.00008
ATM	11	108142837	G	A	rs147575621	NO	missense	ENST00000227616:4:3304G=A	ENSP00000227616:4:p.Gly1102Arg	G1102R	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	9.227	benign	0.001	tolerated	0.55	0	0.00004
ATM	11	108142861	G	A	rs147112946	NO	missense	ENST00000227616:4:3328G=A	ENSP00000227616:4:p.Ala1110Thr	A1110T	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	21.5	benign	0.097	tolerated	0.21	0	0.00007
ATM	11	108142875	G	A	rs138993322	NO	synonymous	ENST00000227616:4:3342G=A	NA	K1114K	PTIR1139;PTIR1139:SF86	9.436	NA	NA	NA	NA	NA	0.00167
ATM	11	108142885	A	G	rs972564322	NO	missense	ENST00000227616:4:3352A=G	ENSP00000227616:4:p.Thr1118Ala	T1118A	PTIR1139;PTIR1139:SF86	11.56	benign	0.001	tolerated	0.36	0	0.00000
ATM	11	108142887	A	G	rs377316692	NO	synonymous	ENST00000227616:4:3354A=G	NA	T1118T	PTIR1139;PTIR1139:SF86	2.459	NA	NA	NA	NA	NA	0.00006
ATM	11	108142892	A	G	rs587780541;rs587781752	NO	synonymous	ENST00000227616:4:3360A=G	NA	A1121A	PTIR1139;PTIR1139:SF86	10.21	NA	NA	NA	NA	NA	0.00000
ATM	11	108150304	A	T	NA	NO	missense	ENST00000227616:4:3371A=T	ENSP00000227616:4:p.Tyr1124Phe	Y1124F	PTIR1139;PTIR1139:SF86	20.5	benign	0.097	deleterious	0.03	15	0.00000
ATM	11	108150311	A	G	rs149182949	NO	synonymous	ENST00000227616:4:3378A=G	NA	K1126K	PTIR1139;PTIR1139:SF86	3.043	NA	NA	NA	NA	NA	0.00018
ATM	11	108150316	A	G	CM021332,rs222902R;COSM21936,COSM1350	NO	missense	ENST00000227616:4:3383A=G	ENSP00000227616:4:p.Gln1128Arg	Q1128R	PTIR1139;PTIR1139:SF86	21.9	benign	0.005	deleterious	0.02	0	0.00000
ATM	11	108150364	TATC	T	rs545376166	NO	intron_feature_truncation	ENST00000227616:4:3402-32_3402+34delCAT	NA	NA	NA	7.454	NA	NA	NA	NA	NA	0.00000
ATM	11	108150372	G	A	rs200168792	NO	intron	ENST00000227616:4:3402-37G=A	NA	NA	NA	4.275	NA	NA	NA	NA	NA	0.00000
ATM	11	108150374	T	C	rs727402941	NO	intron	ENST00000227616:4:3402-39T=C	NA	NA	NA	5.233	NA	NA	NA	NA	NA	0.00000
ATM	11	108150374	T	TA	NA	NO	intron_feature_elongation	ENST00000227616:4:3403-45dupA	NA	NA	NA	1.829	NA	NA	NA	NA	NA	0.00000
ATM	11	108150383	T	A	rs148368017	NO	intron	ENST00000227616:4:3403-34T=A	NA	NA	NA	7.549	NA	NA	NA	NA	NA	0.00000
ATM	11	108150386	T	TA	rs769564716	NO	intron_feature_elongation	ENST00000227616:4:3403-16_3403-15insA	NA	NA	NA	0.011	NA	NA	NA	NA	NA	0.00000
ATM	11	108150726	A	G	rs764890475	NO	missense	ENST00000227616:4:3487A=G	ENSP00000227616:4:p.His1136Arg	H1136R	PTIR1139;PTIR1139:SF86	0.003	benign	0.001	tolerated	0.19	0	0.00000
ATM	11	108150740	C	A	NA	NO	missense	ENST00000227616:4:3421C=A	ENSP00000227616:4:p.Pro1141Thr	P1141T	PTIR1139;PTIR1139:SF86	0.548	benign	0.003	tolerated	0.17	0	0.00000
ATM	11	108150786	T	A	rs759951391;CM910499	NO	missense	ENST00000227616:4:3467C=T	ENSP00000227616:4:p.Thr1156Ala	T1156M	PTIR1139;PTIR1139:SF86	15.37	benign	0.002	tolerated	0.18	0	0.00000
ATM	11	108150787	G	A	rs148358896	NO	synonymous	ENST00000227616:4:3468G=A	NA	T1156T	PTIR1139;PTIR1139:SF86	14.59	NA	NA	NA	NA	NA	0.00012
ATM	11	108150797	G	C	rs545344545	NO	missense	ENST00000227616:4:3478G=C	ENSP00000227616:4:p.Val1160Leu	V1160L	PTIR1139;PTIR1139:SF86	17.79	benign	0.022	tolerated	0.13	0	0.00003
ATM	11	108150836	T	C	rs141460670	NO	synonymous	ENST00000227616:4:3517T=C	NA	L1173L	PTIR1139;PTIR1139:SF86	2.645	NA	NA	NA	NA	NA	0.00006
ATM	11	108150840	T	C	NA	NO	missense	ENST00000227616:4:3521T=C	ENSP00000227616:4:p.Phe1174Ser	F1174S	PTIR1139;PTIR1139:SF86	29.4	probably_damaging	0.993	deleterious	0	65	0.00000
ATM	11	108153392	A	T	NA	NO	intron	ENST00000227616:4:3577-45A=T	NA	NA	NA	6.103	NA	NA	NA	NA	NA	0.00000
ATM	11	108153409	T	C	rs773316929	NO	intron	ENST00000227616:4:3577-28T=C	NA	NA	NA	4.884	NA	NA	NA	NA	NA	0.00000
ATM	11	108153420	C	T	NA	NO	intron	ENST00000227616:4:3577-17C=T	NA	NA	NA	1.359	NA	NA	NA	NA	NA	0.00000
ATM	11	108153431	G	A	rs540806345	NO	splice_intron	ENST00000227616:4:3577-6G=A	NA	NA	NA	0.101	NA	NA	NA	NA	NA	0.00000
ATM	11	108153448	A	G	rs376524625	NO	synonymous	ENST00000227616:4:3588A=G	NA	K1196K	PTIR1139;PTIR1139:SF86	0.089	NA	NA	NA	NA	NA	0.00009
ATM	11	108153466	A	T	NA	NO	synonymous	ENST00000227616:4:3606A=T	NA	G1202G	PTIR1139;PTIR1139:SF86	0.102	NA	NA	NA	NA	NA	0.00000
ATM	11	108153474	G	A	rs749106895	NO	missense	ENST00000227616:4:3614G=A	ENSP00000227616:4:p.Arg1205His	R1205H	PTIR1139;PTIR1139:SF86	7.767	benign	0.001	tolerated	0.26	0	0.00000
ATM	11	108153489	T	C	rs192572042	NO	missense	ENST00000227616:4:3629T=C	ENSP00000227616:4:p.Met1210Thr	M1210T	PTIR1139;PTIR1139:SF86	23.1	benign	0.131	deleterious	0.02	0	0.00000
ATM	11	108153490	G	A	rs587778073;CM023333	NO	missense	ENST00000227616:4:3630G=A	ENSP00000227616:4:p.Met1210Ile	M1210I	PTIR1139;PTIR1139:SF86	21.7	benign	0.017	tolerated	0.16	0	0.00000
ATM	11	108153503	G	C	NA	NO	synonymous	ENST00000227616:4:3643G=C	ENSP00000227616:4:p.Asp1215Glu	D1215H	PTIR1139;PTIR1139:SF86	24.3	probably_damaging	0.921	tolerated	0.19	0	0.00000
ATM	11	108153549	A	G	rs587782195	NO	missense	ENST00000227616:4:3689A=G	ENSP00000227616:4:p.Asn1230Ser	N1230S	PTIR1139;PTIR1139:SF86	0.528	benign	0	tolerated	1	0	0.00000
ATM	11	108153623	C	T	NA	NO	intron	ENST00000227616:4:3746+17C=T	NA	NA	NA	2.428	NA	NA	NA	NA	NA	0.00000
ATM	11	108153654	T	C	NA	NO	intron	ENST00000227616:4:3746+487C=A	NA	NA	NA	4.34	NA	NA	NA	NA	NA	0.00000
ATM	11	108155000	G	A	rs371526361	NO	missense	ENST00000227616:4:3793T=G	ENSP00000227616:4:p.Phe1265Val	F1265V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	24.1	possibly_damaging	0.49	tolerated	0.07	0	0.00000
ATM	11	108155007	AG	A	rs765155119	YES	frameshift_feature_truncation	ENST00000227616:4:3802delA	ENSP00000227616:4:p.Val1268Ser	F1267S	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	28.3	NA	NA	NA	NA	NA	0.00000
ATM	11	108155013	A	G	rs146817595	NO	missense	ENST00000227616:4:3806A=G	ENSP00000227616:4:p.Lys1269Arg	K1269R	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	22	benign	0.033	tolerated	0.06	25	0.00003
ATM	11	108155051	C	A	NA	NO	missense	ENST00000227616:4:3844C=A	ENSP00000227616:4:p.Leu1282Ile	L1282I	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	24.3	benign	0.030	tolerated	0.02	0	0.00000
ATM	11	108155080	C	T	NA	NO	missense	ENST00000227616:4:3883C=T	ENSP00000227616:4:p.Leu1295Phe	L1295F	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	30	probably_damaging	0.997	deleterious	0	15	0.00000
ATM	11	108155132	G	A	rs149711778;COSM22507	NO	missense	ENST00000227616:4:3935G=A	ENSP00000227616:4:p.Ala1309Thr	A1309T	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	20.6	benign	0.027	tolerated	0.08	0	0.00153
ATM	11	108155168	A	G	rs730881366	NO	missense	ENST00000227616:4:3961A=G	ENSP00000227616:4:p.Met1321Val	M1321V	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	7.622	benign	0.001	tolerated	0.6	0	0.00000
ATM	11	108155200	G	C	CO004369131;COSM1350849;COSM1350848	NO	missense_splice	ENST00000227616:4:3993G=C	ENSP00000227616:4:p.Gln1331His	Q1331H	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	25.8	possibly_damaging	0.806	deleterious	0.01	0	0.00000
ATM	11	108155205	G	T	CS023467;rs3092642	NO	splice_intron	ENST00000227616:4:3993-5G=T	NA	NA	NA	0.154	NA	NA	NA	NA	NA	0.00000
ATM	11	108155234	A	G	NA	NO	intron	ENST00000227616:4:3993-38A=G	NA	NA	NA	2.868	NA	NA	NA	NA	NA	0.00000
ATM	11	108155240	G	A	rs2218709	NO	intron	ENST00000227616:4:3993-48G=A	NA	NA	NA	5.965	NA	NA	NA	NA	NA	0.00000
ATM	11	108155248	A	G	rs2218712	NO	intron	ENST00000227616:4:3993-48A=G	NA	NA	NA	1.71	NA	NA	NA	NA	NA	0.00000
ATM	11	108155251	T	C	rs767641380	NO	intron	ENST00000227616:4:3993-51T=C	NA	NA	NA	3.254	NA	NA	NA	NA	NA	0.00000
ATM	11	108155275	T	C	NA	NO	intron	ENST00000227616:4:3993-75T=C	NA	NA	NA	1.067	NA	NA	NA	NA	NA	0.00000
ATM	11	108155279	C	T	rs142218835	NO	intron	ENST00000227616:4:3993-78C=T	NA	NA	NA	0.859	NA	NA	NA	NA	NA	0.00000
ATM	11	108155280	G	A	rs575874976	NO	intron	ENST00000227616:4:3993-80G=A	NA	NA	NA	0.797	NA	NA	NA	NA	NA	0.00000
ATM	11	108155289	G	A	NA	NO	intron	ENST00000227616:4:3993-89G=A	NA	NA	NA	0.568	NA	NA	NA	NA	NA	0.00000
ATM	11	108155297	A	G	NA	NO	intron	ENST00000227616:4:3993-97A=G	NA	NA	NA	3.863	NA	NA	NA	NA	NA	0.00000
ATM	11	108155277	C	T	rs753142634	NO	intron	ENST00000227616:4:3994-98C=T	NA	NA	NA	0.601	NA	NA	NA	NA	NA	0.00000
ATM	11	108155288	A	G	rs778072373	NO	intron	ENST00000227616:4:3994-88A=G	NA	NA	NA	4.549	NA	NA	NA	NA	NA	0.00000
ATM	11	108155289	T	C	NA	NO	intron	ENST00000227616:4:3994-387C=T	NA	NA	NA	5.229	NA	NA	NA	NA	NA	0.00000
ATM	11	108155320	C	T	rs769946400	NO	splice_intron	ENST00000227616:4:3994-7C=T	NA	NA	NA	12.49	NA	NA	NA	NA	NA	0.00000
ATM	11	108155375	T	C	rs56355811	NO	missense	ENST00000227616:4:3942T=C	ENSP00000227616:4:p.Ser1314Pro	L1314H	Superfamily_domain:SSF48371;PTIR1139;PTIR1139:SF86	2.599	NA	NA	NA	NA	NA	0.00006
ATM	11	108155393	C	A

ATM	11	108172464	C	G	NA	NO	missense	ENST00000276146.4:c.526ZC>G	ENSP00000276146.4:p.Tm7356Ag	T7356R	Superfamily_domains:SSF48371,PTHR11139:PTHR11139:SF86	13.1	benign	0.001	tolerated	0.43	0	0.00000
ATM	11	108172486	TC	T	rs587778646,CD991607,COSM21933	YES	frameshift_feature_truncation	ENST00000276146.4:c.529delZC	ENSP00000276146.4:p.Leu1764Tyrfs*Ter12	L1764X	Superfamily_domains:SSF48371,PTHR11139:PTHR11139:SF86	34	NA	NA	NA	NA	NA	0.00003
ATM	11	108172491	A	G	rs555563590	NO	missense	ENST00000276146.4:c.529AA>G	ENSP00000276146.4:p.Glu1765Asp	Q1765R	Superfamily_domains:SSF48371,PTHR11139:PTHR11139:SF86	34.37	benign	0.002	tolerated	0.44	0	0.00000
ATM	11	108172533	A	G	NA	NO	intron	ENST00000276146.4:c.5119+17A>G	NA	NA	NA	9.284	NA	NA	NA	NA	NA	0.00000
ATM	11	108172545	T	C	rs746785712	NO	intron	ENST00000276146.4:c.5119+28T>C	NA	NA	NA	5.968	NA	NA	NA	NA	NA	0.00000
ATM	11	108173547	C	T	rs749105235	NO	intron	ENST00000276146.4:c.5120-33C>T	NA	NA	NA	10.32	NA	NA	NA	NA	NA	0.00000
ATM	11	108173577	T	C	NA	NO	splice_intron	ENST00000276146.4:c.5120-1T>C	NA	NA	NA	12.79	NA	NA	NA	NA	NA	0.00000
ATM	11	108173583	T	G	rs768828004	NO	missense	ENST00000276146.4:c.5121T>G	ENSP00000276146.4:p.Leu1775Val	L1775V	PTHR11139:PTHR11139:SF86	10.59	benign	0.437	tolerated	0.85	0	0.00000
ATM	11	108173612	C	T	rs140641762	NO	synonymous	ENST00000276146.4:c.5152C>T	NA	N1784N	PTHR11139:PTHR11139:SF86	4.862	NA	NA	NA	NA	NA	0.00006
ATM	11	108173663	A	G	NA	NO	missense	ENST00000276146.4:c.5203A>G	ENSP00000276146.4:p.Ser1799Gly	S1799G	PTHR11139:PTHR11139:SF86	1.395	benign	0.001	tolerated	0.43	NA	0.00000
ATM	11	108173665	A	G	NA	NO	missense	ENST00000276146.4:c.5405A>G	ENSP00000276146.4:p.His1802Arg	H1802R	PTHR11139:PTHR11139:SF86	26.2	probably_damaging	0.987	deleterious	0	25	0.00000
ATM	11	108173669	C	T	NA	NO	synonymous	ENST00000276146.4:c.5409C>T	NA	D1803D	PTHR11139:PTHR11139:SF86	7.89	NA	NA	NA	NA	NA	0.00000
ATM	11	108173670	A	T	rs769872474,CM091015[12]	NO	missense	ENST00000276146.4:c.5410A>T	ENSP00000276146.4:p.Ile1804Phe	I1804F	PTHR11139:PTHR11139:SF86	0.06	benign	0.025	tolerated	0.29	0	0.00003
ATM	11	108173730	C	T	rs764784477	NO	missense	ENST00000276146.4:c.5470C>T	ENSP00000276146.4:p.Leu1824Phe	L1824F	PTHR11139:PTHR11139:SF86	24.3	benign	0.112	deleterious	0.01	15	0.00000
ATM	11	108173356	C	T	NA	NO	intron	ENST00000276146.4:c.5497-66C>T	NA	NA	NA	2.423	NA	NA	NA	NA	NA	0.00000
ATM	11	108173387	G	C	rs1002528	NO	intron	ENST00000276146.4:c.5497-13G>C	NA	NA	NA	0.772	NA	NA	NA	NA	NA	0.00000
ATM	11	108173394	T	C	rs1092829	NO	splice_intron	ENST00000276146.4:c.5497-83T>C	NA	NA	NA	3.28	NA	NA	NA	NA	NA	0.00000
ATM	11	108173440	A	C	CS991306	YES	splice_acceptor	ENST00000276146.4:c.5497-2A>C	NA	NA	NA	24	NA	NA	NA	NA	NA	0.00000
ATM	11	108173420	T	C	CM980143	YES	stop_gained	ENST00000276146.4:c.5515C>T	ENSP00000276146.4:p.Gln1839Ter	Q1839*	PTHR11139:PTHR11139:SF86	46	NA	NA	NA	NA	NA	0.00000
ATM	11	108173427	T	NA	NA	NO	intron	ENST00000276146.4:c.5522T>[]	ENSP00000276146.4:p.Val1841Ala	V1841A	PTHR11139:PTHR11139:SF86	19.68	benign	0.045	tolerated	0.14	25	0.00000
ATM	11	108173429	C	T	NA	NO	missense	ENST00000276146.4:c.5524C>T	ENSP00000276146.4:p.Leu1842Phe	L1842F	PTHR11139:PTHR11139:SF86	24.4	possibly_damaging	0.607	tolerated	0.2	0	0.00000
ATM	11	108173455	A	G	rs13850888	NO	synonymous	ENST00000276146.4:c.5550A>G	NA	L1850L	PTHR11139:PTHR11139:SF86	4.996	NA	NA	NA	NA	NA	0.00000
ATM	11	108173456	C	T	rs200842502	NO	missense	ENST00000276146.4:c.5551C>T	ENSP00000276146.4:p.Leu1851Phe	L1851F	PTHR11139:PTHR11139:SF86	22.3	benign	0.045	tolerated	0.12	0	0.00000
ATM	11	108173459	C	T	rs774562056	YES	stop_gained	ENST00000276146.4:c.5554C>T	ENSP00000276146.4:p.Gln1852Ter	Q1852*	PTHR11139:PTHR11139:SF86	42	NA	NA	NA	NA	NA	0.00003
ATM	11	108173462	G	A	CM077896,rs1801516,COSM41596,COSM1738NO	NO	missense	ENST00000276146.4:c.5557G>A	ENSP00000276146.4:p.Ala1853Asn	D1853N	PTHR11139:PTHR11139:SF86	23.2	benign	0.029	tolerated	0.23	0	0.26659
ATM	11	108173463	A	C	rs180435713,CM003593,COSM1350893,COSM21NO	NO	missense	ENST00000276146.4:c.5558A>C	ENSP00000276146.4:p.Asp1853Ala	D1853V	PTHR11139:PTHR11139:SF86	23.6	benign	0.137	deleterious	0	15	0.01344
ATM	11	108173485	T	C	NA	NO	synonymous	ENST00000276146.4:c.5580T>C	ENSP00000276146.4:p.Ser1860Pro	N1860N	PTHR11139:PTHR11139:SF86	2.99	NA	NA	NA	NA	NA	0.00000
ATM	11	108173488	G	A	rs781138087	NO	synonymous	ENST00000276146.4:c.5583G>A	NA	L1861L	PTHR11139:PTHR11139:SF86	7.463	NA	NA	NA	NA	NA	0.00012
ATM	11	108173493	T	C	NA	NO	synonymous	ENST00000276146.4:c.5588T>C	NA	L1862L	PTHR11139:PTHR11139:SF86	0.279	NA	NA	NA	NA	NA	0.00000
ATM	11	108173500	T	C	rs772261410	NO	synonymous	ENST00000276146.4:c.5595T>C	NA	H1865H	PTHR11139:PTHR11139:SF86	0.055	NA	NA	NA	NA	NA	0.00000
ATM	11	108173528	C	T	rs756603775,CM980144,COSM1350897,COSNYES	YES	stop_gained	ENST00000276146.4:c.5623C>T	ENSP00000276146.4:p.Arg1875Ter	R1875*	PTHR11139:PTHR11139:SF86	36	NA	NA	NA	NA	NA	0.00000
ATM	11	108173544	C	T	rs587780628,CM981145212,COSM1145211	NO	missense	ENST00000276146.4:c.5639C>T	ENSP00000276146.4:p.Thr1880Ile	T1880I	PTHR11139:PTHR11139:SF86	12.07	benign	0.011	tolerated	0.06	0	0.00000
ATM	11	108173549	G	T	CM010193	YES	stop_gained	ENST00000276146.4:c.5644G>T	ENSP00000276146.4:p.Glu1882Ter	E1882*	PTHR11139:PTHR11139:SF86	38	NA	NA	NA	NA	NA	0.00000
ATM	11	108173574	A	G	NA	NO	missense	ENST00000276146.4:c.5669A>G	ENSP00000276146.4:p.Asp1890Gly	D1890G	PTHR11139:PTHR11139:SF86	23.7	benign	0.009	deleterious	0.01	0	0.00000
ATM	11	108173610	G	A	rs550070837	NO	intron	ENST00000276146.4:c.5674-31G>A	NA	NA	NA	6.376	NA	NA	NA	NA	NA	0.00000
ATM	11	108173640	A	G	NA	NO	intron	ENST00000276146.4:c.5674-61A>G	NA	NA	NA	3.508	NA	NA	NA	NA	NA	0.00000
ATM	11	108173644	A	C	NA	NO	missense	ENST00000276146.4:c.5712A>C	ENSP00000276146.4:p.Ile1904Ser	K1904N	PTHR11139:PTHR11139:SF86	12.25	benign	0.001	tolerated	0.52	0	0.00000
ATM	11	108173668	A	C	NA	NO	synonymous	ENST00000276146.4:c.5719A>C	NA	R1907R	PTHR11139:PTHR11139:SF86	0.788	NA	NA	NA	NA	NA	0.00000
ATM	11	108173702	G	C	rs148064985	NO	missense	ENST00000276146.4:c.5753G>C	ENSP00000276146.4:p.Arg1918Thr	R1918T	PTHR11139:PTHR11139:SF86	22.1	benign	0.206	tolerated	0.12	0	0.00025
ATM	11	108173032	A	G	NA	NO	intron	ENST00000276146.4:c.5763-55A>G	NA	NA	NA	0.297	NA	NA	NA	NA	NA	0.00000
ATM	11	108180841	T	C	rs776492831	NO	intron	ENST00000276146.4:c.5763-68T>C	NA	NA	NA	5.95	NA	NA	NA	NA	NA	0.00000
ATM	11	108180917	T	C	rs1092910	NO	synonymous	ENST00000276146.4:c.5795T>C	NA	A1931A	PTHR11139:PTHR11139:SF86	7.568	NA	NA	NA	NA	NA	0.01263
ATM	11	108180945	G	C	rs147137706,CM094647,COSM21922	NO	missense	ENST00000276146.4:c.5821G>C	ENSP00000276146.4:p.Val1941Leu	V1941L	PTHR11139:PTHR11139:SF86	24.5	possibly_damaging	0.573	tolerated	0.08	0	0.00030
ATM	11	108181006	A	G	CM094670,rs539931,COSM21918	NO	missense	ENST00000276146.4:c.5882A>G	ENSP00000276146.4:p.Tyr1963Cys	Y1963C	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	28.3	probably_damaging	0.993	deleterious	0	45	0.00012
ATM	11	108181011	G	A	NA	NO	missense	ENST00000276146.4:c.5887G>A	ENSP00000276146.4:p.Ala1963Thr	D1963N	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	25.5	possibly_damaging	0.473	deleterious	0.22	0	0.00000
ATM	11	108181014	A	G	rs201963507,CM085264,CM091015[14	NO	missense	ENST00000276146.4:c.5890A>G	ENSP00000276146.4:p.Ile1964Glu	K1964E	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	17.81	benign	0.025	tolerated	0.45	0	0.00024
ATM	11	108181052	CA	NA	NA	YES	frameshift_feature_truncation	ENST00000276146.4:c.5934delA	ENSP00000276146.4:p.Glu1971Asp[5Ter19	Q1970X	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	35	NA	NA	NA	NA	NA	0.00000
ATM	11	108181048	G	A	NA	NO	splice_intron	ENST00000276146.4:c.5918-6G>A	NA	NA	NA	11.04	NA	NA	NA	NA	NA	0.00000
ATM	11	108181059	T	G	NA	NO	intron	ENST00000276146.4:c.5918-17T>G	NA	NA	NA	15.27	NA	NA	NA	NA	NA	0.00000
ATM	11	108181065	G	T	NA	NO	intron	ENST00000276146.4:c.5918-23G>T	NA	NA	NA	7.388	NA	NA	NA	NA	NA	0.00000
ATM	11	108181117	G	A	NA	NO	splice_acceptor	ENST00000276146.4:c.5919-1C>T	NA	NA	NA	22.4	NA	NA	NA	NA	NA	0.00000
ATM	11	108183140	G	GAACTTATTCAANA	NO	inframe_insertion	ENST00000276146.4:c.5921_5922insAACTTATTCAAA	ENSP00000276146.4:p.Ser1974delinsArgThrTyrSerAs	S1974RTYSN	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	11.89	NA	NA	NA	NA	NA	NA	0.00000
ATM	11	108183151	G	T	rs587778852,CM991607,CM980147,COSM1237YES	YES	stop_gained	ENST00000276146.4:c.5932C>T	ENSP00000276146.4:p.Gln1978Ter	Q1978*	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	53	NA	NA	NA	NA	NA	0.00021
ATM	11	108183172	A	G	NA	NO	missense	ENST00000276146.4:c.5953A>G	ENSP00000276146.4:p.Thr1985Ala	T1985A	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	12.62	benign	0.002	tolerated	0.45	0	0.00000
ATM	11	108183175	A	G	NA	NO	missense	ENST00000276146.4:c.5956A>G	ENSP00000276146.4:p.Ile1986Val	I1986V	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	16.94	benign	0.263	tolerated	0.32	0	0.00000
ATM	11	108183193	AAAAGT	A	rs749040519	NO	frameshift_feature_truncation	ENST00000276146.4:c.5975_9983delTAAAG	ENSP00000276146.4:p.Val1992Asfs*Ter23	K3195E-1993X	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	35	NA	NA	NA	NA	NA	0.00000
ATM	11	108183194	A	C	rs150757822	NO	missense	ENST00000276146.4:c.5975A>C	ENSP00000276146.4:p.Lys1992Thr	K1992T	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	23.6	possibly_damaging	0.439	tolerated	0.08	0	0.00093
ATM	11	108183226	G	C	NA	YES	splice_donor	ENST00000276146.4:c.6006+1G>C	NA	NA	NA	25.6	NA	NA	NA	NA	NA	0.00000
ATM	11	108183233	T	C	rs56019194	NO	splice_intron	ENST00000276146.4:c.6006+8T>C	NA	NA	NA	9.444	NA	NA	NA	NA	NA	0.00000
ATM	11	108184499	A	G	rs172978094	NO	intron	ENST00000276146.4:c.6007-51A>G	NA	NA	NA	6.483	NA	NA	NA	NA	NA	0.00000
ATM	11	108186503	A	T	rs755604182	NO	intron	ENST00000276146.4:c.6007-97A>T	NA	NA	NA	5.922	NA	NA	NA	NA	NA	0.00000
ATM	11	108186563	A	C	rs762001297	NO	missense	ENST00000276146.4:c.6020A>C	ENSP00000276146.4:p.Gln2007Ala	Q2007G	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	28.4	possibly_damaging	0.786	deleterious	0	25	0.00003
ATM	11	108186568	T	C	rs995860999	NO	synonymous	ENST00000276146.4:c.6023T>C	ENSP00000276146.4:p.Tyr2008His	Y2008H	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	27.7	probably_damaging	0.963	deleterious	0	35	0.00006
ATM	11	108186599	A	G	CM094662,CS992462,COSM21934,COSM23NO	NO	missense	ENST00000276146.4:c.6056A>G	ENSP00000276146.4:p.Tyr2019Cys	Y2019C	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	27.2	possibly_damaging	0.823	deleterious	0	65	0.00000
ATM	11	108186610	G	A	rs121121587,CM092585,COSM24635	NO	missense	ENST00000276146.4:c.6067G>A	ENSP00000276146.4:p.Gly2023Arg	G2023R	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	31	probably_damaging	0.973	deleterious	0	25	0.00465
ATM	11	108186621	A	G	CM014793,rs145847515	YES	missense	ENST00000276146.4:c.6080A>G	ENSP00000276146.4:p.Ile2030Val	I2030V	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	11.21	benign	0.001	tolerated	0.46	0	0.00012
ATM	11	108186644	T	C	NA	NO	splice_intron	ENST00000276146.4:c.6095+6T>C	NA	NA	NA	4.128	NA	NA	NA	NA	NA	0.00000
ATM	11	108186653	T	C	rs1212321	NO	intron	ENST00000276146.4:c.6095+15T>C	NA	NA	NA	3.457	NA	NA	NA	NA	NA	0.00000
ATM	11	108186663	G	A	rs769865054	NO	intron	ENST00000276146.4:c.6095+28G>A	NA	NA	NA	2.658	NA	NA	NA	NA	NA	0.00000
ATM	11	108186664	G	A	rs775364739	NO	intron	ENST00000276146.4:c.6095+28G>A	NA	NA	NA	1.713	NA	NA	NA	NA	NA	0.00000
ATM	11	108186683	G	A	NA	NO	intron	ENST00000276146.4:c.6095-45G>A	NA	NA	NA	6.66	NA	NA	NA	NA	NA	0.00000
ATM	11	108186685	A	G	NA	NO	intron	ENST00000276146.4:c.6095-47A>G	NA	NA	NA	7.36	NA	NA	NA	NA	NA	0.00000
ATM	11	108186698	G	A	rs785590586	NO	intron	ENST00000276146.4:c.6096-60G>A	NA	NA	NA	2.998	NA	NA	NA	NA	NA	0.00000
ATM	11	108186708	C	T	NA	NO	intron	ENST00000276146.4:c.6096-38C>G	NA	NA	NA	1.197	NA	NA	NA	NA	NA	0.00000
ATM	11	108186742	C	A	rs1324080 70,CM060102,COSM922732,COSMINO	NO	synonymous	ENST00000276146.4:c.6108C>A	NA	R2034R	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	15.53	NA	NA	NA	NA	NA	0.00003
ATM	11	108186743	G	A	rs121867870,COSM1315815	NO	missense	ENST00000276146.4:c.6109G>A	ENSP00000276146.4:p.Arg2034Gln	R2034Q	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	23.1	benign	0.018	tolerated	0.08	0	0.00000
ATM	11	108186754	C	T	NA	NO	synonymous	ENST00000276146.4:c.6112C>T	ENSP00000276146.4:p.His2038Tyr	H2038Y	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	23.9	benign	0.26	deleterious	0	65	0.00000
ATM	11	108186756	C	T	rs774993357,CS002006670	NO	synonymous	ENST00000276146.4:c.6114C>T	NA	H2038H	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	13.13	NA	NA	NA	NA	NA	0.00000
ATM	11	108186795	C	T	NA	NO	synonymous	ENST00000276146.4:c.6153C>T	NA	L2051L	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	11.84	NA	NA	NA	NA	NA	0.00000
ATM	11	108186807	G	A	rs779646229	NO	synonymous	ENST00000276146.4:c.6165C>A	NA	L2055L	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	10.15	NA	NA	NA	NA	NA	0.00003
ATM	11	108186817	A	G	NA	NO	missense	ENST00000276146.4:c.6175A>G	ENSP00000276146.4:p.Thr2059Ala	T2059A	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	13.99	benign	0.007	tolerated	0.3	0	0.00000
ATM	11	108186820	C	T	rs587778078	NO	synonymous	ENST00000276146.4:c.6178C>T	ENSP00000276146.4:p.Arg2060Cys	R2060C	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	27	benign	0.022	deleterious	0.05	15	0.00006
ATM	11	108186823	C	T	NA	YES	stop_gained	ENST00000276146.4:c.6181C>T	ENSP00000276146.4:p.Gln2061Ter	Q2061*	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	45	NA	NA	NA	NA	NA	0.00000
ATM	11	108186833	T	TC	NA	YES	frameshift_feature_elongation	ENST00000276146.4:c.6192dupC	ENSP00000276146.4:p.Ile2065HisfsTer23	I2064CX	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	35	NA	NA	NA	NA	NA	0.00000
ATM	11	108186884	G	A	NA	NO	intron	ENST00000276146.4:c.6198+44G>A	NA	NA	NA	5.162	NA	NA	NA	NA	NA	0.00000
ATM	11	108186885	C	T	NA	NO	intron	ENST00000276146.4:c.6198+45C>T	NA	NA	NA	7.489	NA	NA	NA	NA	NA	0.00000
ATM	11	108188120	C	T	rs752478345	NO	synonymous	ENST00000276146.4:c.6213A>G	NA	L2071L	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	14.86	NA	NA	NA	NA	NA	0.00000
ATM	11	108188135	C	T	rs769483748	NO	synonymous	ENST00000276146.4:c.6234C>T	NA	S2078S	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	12.14	NA	NA	NA	NA	NA	0.00018
ATM	11	108188136	G	A	rs1800060	NO	synonymous	ENST00000276146.4:c.6235A>G	ENSP00000276146.4:p.Val2079Ile	V2079I	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	0.005	benign	0.001	tolerated	NA	0	0.00243
ATM	11	108188149	G	C	NA	NO	missense	ENST00000276146.4:c.6248G>C	ENSP00000276146.4:p.Gly2083Ala	G2083A	PTHR11139:PTHR11139:SF86,PROSITE_profiles:PS51189	22.5	benign	0.405	tolerated	0.07	55	0.00000
ATM	11	108188194	T	C	rs587780631	NO	missense	ENST00000276146.4:c.6293T>C	ENSP00000276146.4:p.Leu2098Pro	L2098P	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	23.4	benign	0.217	deleterious	0.02	0	0.00003
ATM	11	108188233	G	C	CM015635	NO	missense	ENST00000276146.4:c.6332A>G	ENSP00000276146.4:p.His2111Arg	H2111R	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	4.286	benign	0.027	tolerated	0.35	0	0.00000
ATM	11	108188234	T	C	rs575576349	NO	synonymous	ENST00000276146.4:c.6333T>C	NA	H2111H	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	0.011	NA	NA	NA	NA	NA	0.00000
ATM	11	108188267	T	C	rs766810400	NO	intron	ENST00000276146.4:c.6347-19T>C	NA	NA	NA	11.65	NA	NA	NA	NA	NA	0.00000
ATM	11	108188627	T	C	NA	NO	regulatory	ENST00000276146.4:c.6544-56T>C	NA	NA	NA	1.898	NA	NA	NA	NA	NA	0.00000
ATM	11	108188627	T	C	rs116924981	NO	regulatory	ENST00000276146.4:c.6544-54T>C	NA	NA	NA	2.495	NA	NA	NA	NA	NA	0.00000
ATM	11	108188633	T	G	NA	NO	regulatory	ENST00000276146.4:c.6548-48T>G	NA	NA	NA	4.484	NA	NA	NA	NA	NA	0.00000
ATM	11	108188638	G	A	NA	NO	regulatory	ENST00000276146.4:c.6548-43G>A	NA	NA	NA	5.288	NA	NA	NA	NA	NA	0.00000
ATM	11	108188652	T	G	rs771558112	NO	regulatory	ENST00000276146.4:c.6548-29T>G	NA	NA	NA	7.248	NA	NA	NA	NA	NA	0.00000
ATM	11	108188673	T	C	rs730881292	NO	splice_intron	ENST00000276146.4:c.6548-87T>C	NA	NA	NA	0.679	NA	NA	NA	NA	NA	0.00000
ATM	11	108188703	A	C	NA	NO	missense	ENST00000276146.4:c.6587G>C	ENSP00000276146.4:p.Tyr2124His	Y2124H	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	27.1	probably_damaging	0.996	deleterious	0	25	0.00000
ATM	11	108188727	C	T	rs551408889,COSM336196,COSM1368195	NO	synonymous	ENST00000276146.4:c.6594C>T	NA	L2132L	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	13.42	NA	NA	NA	NA	NA	0.00003
ATM	11	108188736	G	CTT	rs587782554,CS962208	YES	frameshift_feature_elongation	ENST00000276146.4:c.6604_6605insTT	ENSP00000276146.4:p.Ser2135Ter	L2135LX	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	35	NA	NA	NA	NA	NA	0.00000
ATM	11	108188764	A	G	NA	NO	missense	ENST00000276146.4:c.6631A>G	ENSP00000276146.4:p.Tyr2144Cys	Y2144C	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	23.7	possibly_damaging	0.762	tolerated	0.17	15	0.00000
ATM	11	108188770	G	A	rs5685 5849	NO	missense	ENST00000276146.4:c.6637G>A	ENSP00000276146.4:p.Ser2146Thr	S2146T	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	7.386	benign	0.002	tolerated	1	0	0.00006
ATM	11	108188810	G	A	rs763680797	NO	intron	ENST00000276146.4:c.6652-25G>A	NA	NA	NA	4.483	NA	NA	NA	NA	NA	0.00000
ATM	11	108188810	C	T	rs763680797	NO	intron	ENST00000276146.4:c.6652-25C>T	NA	NA	NA	1.62	NA	NA	NA	NA	NA	0.00000
ATM	11	108188847	A	G	NA	NO	intron	ENST00000276146.4:c.6652+62A>G	NA	NA	NA	5.511	NA	NA	NA	NA	NA	0.00000
ATM	11	108189194	T	C	NA	NO	intron	ENST00000276146.4:c.6653-347T>C	NA	NA	NA	1.696	NA	NA	NA	NA	NA	0.00000
ATM	11	108192080	CAG	C	rs745661253	NO	intron_feature_truncation	ENST00000276146.4:c.6655-25_6453-24delAG	NA	NA	NA	7.915	NA	NA	NA	NA	NA	0.00000
ATM	11	108192080	G	A	rs140622883	NO	synonymous	ENST00000276146.4:c.6663G>A	ENSP00000276146.4:p.Val2221Val	V2221V	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	11.8	NA	NA	NA	NA	NA	0.00000
ATM	11	108192507	G	A	rs756626462	NO	missense	ENST00000276146.4:c.6820G>A	ENSP00000276146.4:p.Arg2241His	R2241H	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	24.2	benign	0.038	tolerated	0.08	0	0.00000
ATM	11	108192078	C	T	rs200431631,COSM1344164,COSM3443163	NO	missense	ENST00000276146.4:c.6830C>T	ENSP00000276146.4:p.Ser2244Leu	S2244L	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	29.5	possibly_damaging	0.763	tolerated	0.06	0	0.00000
ATM	11	108192082	G	A	NA	NO	synonymous	ENST00000276146.4:c.6834G>A	ENSP00000276146.4:p.Leu2245Leu	L2245L	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	9.912	NA	NA	NA	NA	NA	0.00000
ATM	11	108192101	T	C	rs143715818	NO	synonymous	ENST00000276146.4:c.6852G>C	ENSP00000276146.4:p.Leu2284Leu	L2284L	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	1.683	NA	NA	NA	NA	NA	0.00000
ATM	11	108192106	G	A	NA	NO	synonymous	ENST00000276146.4:c.6855G>A	ENSP00000276146.4:p.Ser2285Ser	S2285S	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	8.668	NA	NA	NA	NA	NA	0.00000
ATM	11	108192126	G	C	rs774551964	NO	missense	ENST00000276146.4:c.6851G>C	ENSP00000276146.4:p.Ser2184Thr	S2184T	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	14.7	benign	0.102	tolerated	0.09	0	0.00003
ATM	11	108192129	T	C	rs779611511	NO	missense	ENST00000276146.4:c.6854T>C	ENSP00000276146.4:p.Ile2185Thr	I2185T	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	22.8	benign	0.015	tolerated	0.05	0	0.00000
ATM	11	108192158	C	T	rs368049107	NO	intron	ENST00000276146.4:c.6872-11C>T	NA	NA	NA	4.731	NA	NA	NA	NA	NA	0.00000
ATM	11	108192159	G	A	rs5218677	NO	intron	ENST00000276146.4:c.6872+12G>T	NA	NA	NA	0.792	NA	NA	NA	NA	NA	0.00000
ATM	11	108192668	T	G	rs730881311	NO	missense	ENST00000276146.4:c.6904T>G	ENSP00000276146.4:p.Tyr2202Asp	Y2202D	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	25.3	possibly_damaging	0.88	tolerated	0.08	35	0.00003
ATM	11	108192674	A	G	NA	NO	missense	ENST00000276146.4:c.6910A>G	ENSP00000276146.4:p.Ile2204Val	K2204E	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	23.1	benign	0.38	tolerated	0.18	0	0.00000
ATM	11	108192134	A	G	rs45873723	NO	synonymous	ENST00000276146.4:c.6670A>G	ENSP00000276146.4:p.Met2224Val	M2224V	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	23	benign	0.439	deleterious	0.04	0	0.00000
ATM	11	108192141	C	T	rs944632250,CM990217,COSM1644415,COSNNO	NO	missense	ENST00000276146.4:c.6679C>T	ENSP00000276146.4:p.Arg2227Cys	R2227C	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	35	probably_damaging	1	deleterious	0	65	0.00000
ATM	11	108192164	C	T	rs760602228	NO	synonymous	ENST00000276146.4:c.6708C>T	NA	L2234L	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	12.59	NA	NA	NA	NA	NA	0.00006
ATM	11	108192174	C	T	CM002481,rs151181409	NO	missense	ENST00000276146.4:c.6710A>C	ENSP00000276146.4:p.Lys2237Thr	K2237T	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	23.4	possibly_damaging	0.716	tolerated	0.11	0	0.00000
ATM	11	108192203	A	G	rs587781521	NO	missense	ENST00000276146.4:c.6739A>G	ENSP00000276146.4:p.Ile2247Val	I2247V	Plam_domain:PF02259:PTHR11139,PTHR11139:SF86,PROSITE_profiles:PS51189	6.863	benign	0.038	tolerated	0.39	0	0.00000
ATM	11	108192259	T	C	rs121868091	NO	synonymous	EN

ATM	11	108286683	TATA-AG	T	rs770881294	YES	frameshift,splice,feature_truncation	ENST00000278616.4:c.8264_8266delATA-AG	ENSP00000278616.4:p.Tyr2755Cysfs*Ter12	YK2755-2756X	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:36	NA	NA	NA	NA	NA	0.00000
ATM	11	108286686	A	T	rs771638537,CM960108,COSMT58333	YES	stop_gained,splice	ENST00000278616.4:c.8266A>T	ENSP00000278616.4:p.Lys2756Ter	K2756*	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:54	NA	NA	NA	NA	NA	0.00003
ATM	11	108286694	T	A	rs747153940	NO	splice_intron	ENST00000278616.4:c.8268+6T>A	NA	NA	NA	6.482	NA	NA	NA	NA	0.00000
ATM	11	108213955	A	G	rs114320959	NO	intron	ENST00000278616.4:c.8269-14A>T	NA	NA	NA	3.089	NA	NA	NA	NA	0.00000
ATM	11	108213949	G	A	rs761623530	NO	missense,splice	ENST00000278616.4:c.8269G>A	ENSP00000278616.4:p.Val2757Met	V2757M	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:24,9	probably_damaging	0.954	tolerated	0.05	15	0.00000
ATM	11	108213952	G	T	NA	NO	missense	ENST00000278616.4:c.8272G>T	ENSP00000278616.4:p.Val2758Phe	V2758F	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:29,2	probably_damaging	0.977	deleterious	0	0	0.00000
ATM	11	108213967	C	T	CM880077,COSMI103943,COSMI103942	YES	stop_gained	ENST00000278616.4:c.8287C>T	ENSP00000278616.4:p.Arg2763Ter	R2763*	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:49	NA	NA	NA	NA	NA	0.00000
ATM	11	108213987	G	A	rs778250035,CM960109,COSMI102564	YES	stop_gained	ENST00000278616.4:c.8307G>A	ENSP00000278616.4:p.Trp2769Ter	W2769*	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:51	NA	NA	NA	NA	NA	0.00000
ATM	11	108214001	TC	T	NA	YES	frameshift,feature_truncation	ENST00000278616.4:c.8325delC	ENSP00000278616.4:p.Ile2774LeufsTer30	V2774X	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:35	NA	NA	NA	NA	NA	0.00000
ATM	11	108214007	T	C	rs764675628	NO	missense	ENST00000278616.4:c.8327T>C	ENSP00000278616.4:p.Ile2776Thr	I2776T	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:27	probably_damaging	0.905	deleterious	0	25	0.00000
ATM	11	108214008	GGTGA	G	rs769139997	YES	splice_donor,intron,feature_truncation	ENST00000278616.4:c.8418+5_8418+8delGTGA	NA	NA	NA	27.3	NA	NA	NA	NA	0.00003
ATM	11	108214132	G	A	rs764464003	NO	intron	ENST00000278616.4:c.8418-14G>A	NA	NA	NA	1.267	NA	NA	NA	NA	0.00000
ATM	11	108216545	C	T	rs877798872,CM990119,COSMI22488,COSMI3NO	NO	missense	ENST00000278616.4:c.8494C>T	ENSP00000278616.4:p.Arg2832Cys	R2832C	Superfamily_domains:SSF56112,SMART_domains:SM00146,Pfam_domain:PF00454,PTHR11139:,35	probably_damaging	0.993	deleterious	0	45	0.00003
ATM	11	108216556	G	A	rs728296539	NO	missense	ENST00000278616.4:c.8504G>A	ENSP00000278616.4:p.Arg2835His	R2835H	Superfamily_domains:SSF56112,SMART_domains:SM00146,Pfam_domain:PF00454,PTHR11139:,35	benign	0.183	tolerated	0.36	0	0.00015
ATM	11	108216597	G	A	rs877832202,CM990210	NO	missense	ENST00000278616.4:c.8546G>A	ENSP00000278616.4:p.Arg2849Gln	R2849Q	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:35	probably_damaging	0.998	deleterious	0	35	0.00000
ATM	11	108216609	C	T	rs141537416,CM031519905,COSMI022753,CONO	NO	synonymous	ENST00000278616.4:c.8559C>T	ENSP00000278616.4:p.Thr2853=	T2853=	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:34	probably_damaging	0.999	deleterious	0	65	0.00015
ATM	11	108216610	G	A	rs140855832	NO	synonymous	ENST00000278616.4:c.8559G>A	ENSP00000278616.4:p.Thr2853=	T2853=	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:34	NA	NA	NA	NA	NA	0.00000
ATM	11	108216617	C	T	rs201958469,CM992588	NO	missense	ENST00000278616.4:c.8566C>T	ENSP00000278616.4:p.Arg2856Cys	R2856C	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:35	probably_damaging	0.995	deleterious	0	25	0.00019
ATM	11	108216623	ACTT	A	NA	NO	inframe_deletion	ENST00000278616.4:c.8578_8580delTTCT	ENSP00000278616.4:p.Ser2860del	TS2858-2859T	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:22,6	NA	NA	NA	NA	NA	0.00000
ATM	11	108216625	T	C	NA	NO	synonymous	ENST00000278616.4:c.8580T>C	NA	T2858T	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:42	NA	NA	NA	NA	NA	0.00000
ATM	11	108216628	T	C	CM981351013,COSMI131032	NO	synonymous	ENST00000278616.4:c.8577C>C	NA	S2859S	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:42,414	NA	NA	NA	NA	NA	0.00000
ATM	11	108216639	A	G	NA	NO	splice_intron	ENST00000278616.4:c.8544+6A>G	NA	NA	NA	15.2	NA	NA	NA	NA	0.00000
ATM	11	108218002	C	A	NA	NO	splice_intron	ENST00000278616.4:c.8585-4C>A	NA	NA	NA	7.693	NA	NA	NA	NA	0.00000
ATM	11	108218013	C	T	rs945276370	NO	synonymous	ENST00000278616.4:c.8592C>T	NA	V2864V	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:40	NA	NA	NA	NA	NA	0.00140
ATM	11	108218017	C	G	rs368666328,CM892275A,COSMI1585328	NO	missense	ENST00000278616.4:c.8596C>G	ENSP00000278616.4:p.Leu2866Val	L2866V	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:16,67	benign	0.371	tolerated	0.84	0	0.00000
ATM	11	108218020	G	T	CM960111	YES	stop_gained	ENST00000278616.4:c.8599G>T	ENSP00000278616.4:p.Gly2867Ter	G2867*	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:45	NA	NA	NA	NA	NA	0.00000
ATM	11	108218101	T	G	rs200198537	NO	intron	ENST00000278616.4:c.8671+9T>G	NA	NA	NA	5.593	NA	NA	NA	NA	0.00000
ATM	11	108218126	A	G	NA	NO	intron	ENST00000278616.4:c.8671-34A>G	NA	NA	NA	16.68	NA	NA	NA	NA	0.00000
ATM	11	108224013	A	G	rs761306969	NO	intron	ENST00000278616.4:c.8672-68A>G	NA	NA	NA	2.49	NA	NA	NA	NA	0.00000
ATM	11	108224493	G	A	rs748192003,CM891052Y,COSMI758347	NO	missense,splice	ENST00000278616.4:c.8672G>A	ENSP00000278616.4:p.Gly2891Asp	G2891D	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:32	probably_damaging	1	deleterious	0	65	0.00000
ATM	11	108224527	T	C	NA	NO	synonymous	ENST00000278616.4:c.8706T>C	NA	T2902T	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:47,465	NA	NA	NA	NA	NA	0.00000
ATM	11	108224555	A	G	rs736676328,CM814034	NO	missense	ENST00000278616.4:c.8734A>G	ENSP00000278616.4:p.Arg2912Gly	R2912G	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:29,1	probably_damaging	0.999	deleterious	0	65	0.00099
ATM	11	108224558	G	T	rs756699044	NO	missense	ENST00000278616.4:c.8737G>T	ENSP00000278616.4:p.Asp2913Tyr	D2913Y	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:33	probably_damaging	1	deleterious	0	65	0.00000
ATM	11	108224593	A	G	NA	NO	synonymous	ENST00000278616.4:c.8772A>	NA	E2924E	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:44,984	NA	NA	NA	NA	NA	0.00000
ATM	11	108224608	G	A	CS981484,CS8M23633,COSMI1351847,COSM3YES	YES	splice_donor	ENST00000278616.4:c.8786+1G>A	NA	NA	NA	26.3	NA	NA	NA	NA	0.00000
ATM	11	108224615	A	C	rs499683839	NO	splice,intron	ENST00000278616.4:c.8786+8A>C	NA	NA	NA	1.963	NA	NA	NA	NA	0.00000
ATM	11	108224620	T	A	NA	NO	intron	ENST00000278616.4:c.8786+15G>A	NA	NA	NA	5.042	NA	NA	NA	NA	0.00000
ATM	11	108224632	G	A	rs764861531	NO	intron	ENST00000278616.4:c.8786+28G>A	NA	NA	NA	5.642	NA	NA	NA	NA	0.00000
ATM	11	108224639	T	C	NA	NO	intron	ENST00000278616.4:c.8786+32T>C	NA	NA	NA	1.47	NA	NA	NA	NA	0.00000
ATM	11	108224644	G	A	NA	NO	intron	ENST00000278616.4:c.8786+37G>A	NA	NA	NA	5.258	NA	NA	NA	NA	0.00000
ATM	11	108225473	A	G	rs139490310	NO	intron	ENST00000278616.4:c.8787-6A>G	NA	NA	NA	2.938	NA	NA	NA	NA	0.00000
ATM	11	108225477	A	AAGAT	rs775653463	NO	intron,feature_elongation	ENST00000278616.4:c.8787-59_8787-56dupGATA	NA	NA	NA	0.933	NA	NA	NA	NA	0.00000
ATM	11	108225483	C	T	rs464493[2]	NO	intron	ENST00000278616.4:c.8787-52C>T	NA	NA	NA	2.77	NA	NA	NA	NA	0.00000
ATM	11	108225489	CA	C	NA	NO	intron,feature_truncation	ENST00000278616.4:c.8787-47delA	NA	NA	NA	8.501	NA	NA	NA	NA	0.00000
ATM	11	108225492	C	T	NA	NO	intron	ENST00000278616.4:c.8787-44C>T	NA	NA	NA	5.115	NA	NA	NA	NA	0.00000
ATM	11	108225493	A	T	rs715181707	NO	intron	ENST00000278616.4:c.8787-43A>T	NA	NA	NA	1.734	NA	NA	NA	NA	0.00000
ATM	11	108225494	T	C	NA	NO	intron	ENST00000278616.4:c.8787-44T>C	NA	NA	NA	1.616	NA	NA	NA	NA	0.00000
ATM	11	108225512	C	T	rs773681524	NO	intron	ENST00000278616.4:c.8787-26C>T	NA	NA	NA	1.293	NA	NA	NA	NA	0.00000
ATM	11	108225605	A	C	rs877782135	NO	splice,intron	ENST00000278616.4:c.8788+6A>C	NA	NA	NA	13.26	NA	NA	NA	NA	0.00000
ATM	11	108225615	T	C	NA	NO	intron	ENST00000278616.4:c.8850+14T>C	NA	NA	NA	11.08	NA	NA	NA	NA	0.00000
ATM	11	108225642	C	T	rs759513987	NO	intron	ENST00000278616.4:c.8850+41C>T	NA	NA	NA	7.912	NA	NA	NA	NA	0.00000
ATM	11	108225651	T	G	rs759954606	NO	intron	ENST00000278616.4:c.8850+55T>G	NA	NA	NA	6.452	NA	NA	NA	NA	0.00000
ATM	11	108225658	C	G	rs765358242	NO	intron	ENST00000278616.4:c.8850+62C>G	NA	NA	NA	2.57	NA	NA	NA	NA	0.00000
ATM	11	108225659	T	C	rs745676907	NO	intron	ENST00000278616.4:c.8850+58T>C	NA	NA	NA	5.544	NA	NA	NA	NA	0.00000
ATM	11	108225661	A	G	rs864143	NO	intron	ENST00000278616.4:c.8850-68A>G	NA	NA	NA	0.543	NA	NA	NA	NA	0.00000
ATM	11	108225787	G	A	NA	NO	intron	ENST00000278616.4:c.8851-62G>A	NA	NA	NA	7.552	NA	NA	NA	NA	0.00000
ATM	11	108225730	T	A	NA	NO	intron	ENST00000278616.4:c.8851-19T>A	NA	NA	NA	7.225	NA	NA	NA	NA	0.00000
ATM	11	108225751	C	A	NA	NO	intron	ENST00000278616.4:c.8851-58C>A	NA	NA	NA	4.712	NA	NA	NA	NA	0.00000
ATM	11	108225758	T	G	NA	NO	intron	ENST00000278616.4:c.8851-51T>G	NA	NA	NA	2.5	NA	NA	NA	NA	0.00000
ATM	11	108225761	G	A	rs778614061	NO	intron	ENST00000278616.4:c.8851-48G>A	NA	NA	NA	3.579	NA	NA	NA	NA	0.00000
ATM	11	108225776	T	G	rs764011368	NO	intron	ENST00000278616.4:c.8851-33T>G	NA	NA	NA	4.866	NA	NA	NA	NA	0.00000
ATM	11	108225806	T	C	rs748874219	NO	splice,intron	ENST00000278616.4:c.8851-3T>C	NA	NA	NA	12.98	NA	NA	NA	NA	0.00000
ATM	11	108225818	T	C	rs776619067,COSM1645451,COSM1351049,CINO	NO	missense	ENST00000278616.4:c.8860T>C	ENSP00000278616.4:p.Tyr2954His	Y2954H	Superfamily_domains:SSF56112,SMART_domains:SM00146,3.30.1010.10,Pfam_domain:PF00454:28	probably_damaging	0.954	deleterious	0	65	0.00003
ATM	11	108225886	T	G	NA	NO	synonymous	ENST00000278616.4:c.8925T>G	ENSP00000278616.4:p.Asp2975Glu=	D2975E	SMART_domains:SM00146,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290:,47	possibly_damaging	0.459	tolerated	0.09	0	0.00000
ATM	11	108225955	C	T	rs770644163,CM960113,COSM428365,COSMYES	YES	stop_gained	ENST00000278616.4:c.8977C>T	ENSP00000278616.4:p.Arg2993Ter	R2993*	SMART_domains:SM00146,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290:	NA	NA	NA	NA	NA	0.00000
ATM	11	108225957	A	G	NA	NO	synonymous	ENST00000278616.4:c.8979A>G	NA	R2993R	SMART_domains:SM00146,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290:	7.387	NA	NA	NA	NA	0.00000
ATM	11	108225961	C	T	rs142322668	NO	missense	ENST00000278616.4:c.8983C>T	ENSP00000278616.4:p.Leu2995Phe	L2995F	SMART_domains:SM00146,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290:	16.24	benign	0.187	tolerated	0.57	0	0.00000
ATM	11	108225974	A	G	NA	NO	intron	ENST00000278616.4:c.8987-10A>G	NA	NA	NA	1.668	NA	NA	NA	NA	0.00000
ATM	11	108225995	A	T	NA	NO	intron	ENST00000278616.4:c.8987-58A>T	NA	NA	NA	1.806	NA	NA	NA	NA	0.00000
ATM	11	108226010	A	C	rs758862688	NO	intron	ENST00000278616.4:c.8988-42A>C	NA	NA	NA	2.138	NA	NA	NA	NA	0.00000
ATM	11	108226086	C	T	CM800656,rs587782292,COSM1560110,COSMINO	NO	missense	ENST00000278616.4:c.9022C>T	ENSP00000278616.4:p.Arg3008Cys	R3008C	Coiled-coils_(Ncoils):Coil,SMART_domains:SM00146,1.10.1070.11,PTHR11139:PTHR11139:SF66:54	probably_damaging	0.988	deleterious	0	25	0.00003
ATM	11	108226109	G	A	NA	NO	synonymous	ENST00000278616.4:c.9045G>A	NA	E3015E	Coiled-coils_(Ncoils):Coil,SMART_domains:SM00146,1.10.1070.11,PTHR11139:PTHR11139:SF66:945	NA	NA	NA	NA	NA	0.00000
ATM	11	108226135	C	T	NA	NO	missense	ENST00000278616.4:c.9071C>T	ENSP00000278616.4:p.Thr3024Ile	T3024I	Coiled-coils_(Ncoils):Coil,1.10.1070.11,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50292:,7	benign	0.098	tolerated	0.27	0	0.00000
ATM	11	108226150	G	A	rs201109629	NO	missense	ENST00000278616.4:c.9086G>A	ENSP00000278616.4:p.Gly3029Asp	G3029D	1.10.1070.11,Pfam_domain:PF02260,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50292:,51.93	benign	0.023	tolerated	0.11	0	0.00051
ATM	11	108226183	T	A	rs769870357	NO	missense	ENST00000278616.4:c.9119T>A	ENSP00000278616.4:p.Ile3040Lys	I3040K	1.10.1070.11,Pfam_domain:PF02260,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290:PS4.3	benign	0.038	deleterious	0.02	0	0.00000
ATM	11	108226187	T	A	NA	NO	synonymous	ENST00000278616.4:c.9123C>T	ENSP00000278616.4:p.Pro3041=	D3041D	1.10.1070.11,Pfam_domain:PF02260,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290:PS4.23	NA	NA	NA	NA	NA	0.00000
ATM	11	108226203	C	T	CM960114,rs121434219,COSM21624,COSM11YES	YES	stop_gained	ENST00000278616.4:c.9139C>T	ENSP00000278616.4:p.Arg3047Ter	R3047*	Pfam_domain:PF02260,PTHR11139:PTHR11139:SF66,PROSITE_profiles:PS50290,PROSITE_pm-49	NA	NA	NA	NA	NA	0.00000
CHR22	22	20038467	G	A	rs778864403	NO	5_prime_UTR	ENST00000238154.6:c.-5C>T	NA	NA	NA	6.344	NA	NA	NA	NA	0.00000
CHR22	22	20038878	A	G	rs121908710	NO	5_prime_UTR	ENST00000238154.6:c.-72C>T	NA	NA	NA	6.767	NA	NA	NA	NA	0.00000
CHR22	22	20038880	G	A	rs877781367	NO	5_prime_UTR	ENST00000238154.6:c.-74C>T	NA	NA	NA	0.888	NA	NA	NA	NA	0.00000
CHR22	22	20038894	A	G	rs773678297	NO	synonymous	ENST00000238154.6:c.162T>C	NA	A54LA	NA	0.483	NA	NA	NA	NA	0.00000
CHR22	22	20039034	C	T	rs758555487	NO	synonymous	ENST00000238154.6:c.159G>A	NA	E53LE	PTHR12444:SF21,PTHR12444	10.19	NA	NA	NA	NA	0.00000
CHR22	22	20039042	C	T	NA	NO	missense	ENST00000238154.6:c.150G>A	ENSP00000229178.6:p.Gly5P4Ser	G52P6	PTHR12444:SF21,PTHR12444	10.95	benign	0.004	tolerated	0.4	0.00000
CHR22	22	20039061	C	A	rs877780180,CM013227286	NO	missense	ENST00000238154.6:c.135G>T	ENSP00000229178.6:p.Arg5?Ile,ua	R51IL	PTHR12444:SF21,PTHR12444	24.9	benign	0.33	deleterious	0.01	0	0.00054
CHR22	22	20039064	C	T	rs877780180,CM013227286	NO	missense	ENST00000238154.6:c.-135G>A	ENSP00000229178.6:p.Arg5?Gln	R51QQ	PTHR12444:SF21,PTHR12444	23.1	benign	0.036	tolerated	0.09	0.00000
CHR22	22	20085107	T	C	rs749728970	NO	intron	ENST00000238154.6:c.1542+18A>G	NA	NA	NA	1.213	NA	NA	NA	NA	0.00000
CHR22	22	20085112	A	T	rs1781716	NO	intron	ENST00000238154.6:c.1542+11T>A	NA	NA	NA	3.48	NA	NA	NA	NA	0.00000
CHR22	22	20085139	G	A	rs877782541	NO	synonymous	ENST00000238154.6:c.1528C>T	ENSP00000229178.6:p.Pro509Leu	P509L	PTHR12444:SF21,PTHR12444	10.4	benign	0.003	tolerated	0.1	0	0.00000
CHR22	22	20090098	G	C	NA	NO	missense	ENST00000238154.6:c.1363C>G	ENSP00000229178.6:p.Arg461Gln	D461E	Low_complexity_(Seg):seg,PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.1.10.110:122,1	benign	0.215	tolerated	0.05	35	0.00000
CHR22	22	20090107	G	C	rs7762749	NO	intron	ENST00000238154.6:c.1375-78C>G	NA	NA	NA	5.976	NA	NA	NA	NA	0.00000
CHR22	22	20090168	T	C	NA	NO	intron	ENST00000238154.6:c.1375+75A>G	NA	NA	NA	17.58	NA	NA	NA	NA	0.00000
CHR22	22	20090189	A	G	rs727418829	NO	intron	ENST00000238154.6:c.1375-287C>T	NA	NA	NA	17.54	NA	NA	NA	NA	0.00000
CHR22	22	20090198	C	T	NA	NO	intron	ENST00000238154.6:c.1375+1TG>A	NA	NA	NA	19.08	NA	NA	NA	NA	0.00000
CHR22	22	20090127	C	T	NA	NO	missense	ENST00000238154.6:c.1363G>A	ENSP00000229178.6:p.Val455Ile	V455I	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:322,8	benign	0.048	tolerated	0.28	25	0.00000
CHR22	22	20090142	C	T	NA	NO	missense	ENST00000238154.6:c.1348G>A	ENSP00000229178.6:p.Glu450Lys	E450K	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:320,6	benign	0.001	tolerated	0.69	0	0.00000
CHR22	22	20090147	A	C	rs17886163	NO	missense	ENST00000238154.6:c.1344T>G	ENSP00000229178.6:p.Ile448Ser	I448S	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:320,3	benign	0.309	tolerated	0.76	15	0.00000
CHR22	22	20090172	T	G	rs877780176	NO	missense	ENST00000238154.6:c.1318A>C	ENSP00000229178.6:p.Ile440Leu	I440L	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:321	probably_damaging	0.909	deleterious	0.01	0	0.00000
CHR22	22	20090175	G	A	rs778989252	YES	stop_gained	ENST00000238154.6:c.1315C>T	ENSP00000229178.6:p.Gln439Ter	Q439*	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S546	NA	NA	NA	NA	NA	0.00000
CHR22	22	20090178	C	A	rs800050801,CM077522	NO	missense	ENST00000238154.6:c.1312G>T	ENSP00000229178.6:p.Asp438Tyr	D438Y	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:321	probably_damaging	0.915	deleterious	0	25	0.00069
CHR22	22	20090207	G	A	CM051021,rs137853011,COSM2935967	NO	missense	ENST00000238154.6:c.1263C>T	ENSP00000229178.6:p.Ser42HPhe	S42HF	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:320,9	probably_damaging	0.989	deleterious	0.01	15	0.00011
CHR22	22	20090220	A	G	rs139266548	NO	missense	ENST00000238154.6:c.1237T>C	ENSP00000229178.6:p.Tyr42Ser	Y42S	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S28,7	probably_damaging	1	deleterious	0.03	65	0.00100
CHR22	22	20090225	C	T	rs149755590	NO	missense	ENST00000238154.6:c.1236G>A	ENSP00000229178.6:p.Ser422Asn	S422N	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S26	possibly_damaging	0.66	tolerated	0.06	0	0.00000
CHR22	22	20090226	TA	T	rs877780174,CD110759	YES	frameshift,feature_truncation	ENST00000238154.6:c.1234delT	ENSP00000229178.6:p.Ser42ValfsTer5	L421X	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S25	NA	NA	NA	NA	NA	0.00016
CHR22	22	20090162	T	C	rs763616647	NO	intron	ENST00000238154.6:c.1259+18A>G	NA	NA	NA	1.475	NA	NA	NA	NA	0.00000
CHR22	22	20090164	A	G	rs780245742	NO	intron	ENST00000238154.6:c.1259-147>A	NA	NA	NA	7.321	NA	NA	NA	NA	0.00000
CHR22	22	20090169	T	C	rs368388249	NO	splice,intron	ENST00000238154.6:c.1259+8A>G	NA	NA	NA	2.738	NA	NA	NA	NA	0.00000
CHR22	22	20090174	T	A	NA	NO	missense	ENST00000238154.6:c.1243G>A	ENSP00000229178.6:p.Val415Ile	V415I	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S13	probably_damaging	0.977	deleterious	0.01	25	0.00000
CHR22	22	20090732	C	A	rs766419039	NO	missense	ENST00000238154.6:c.1225G>T	ENSP00000229178.6:p.Asp409Tyr	D409Y	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S34	probably_damaging	1	deleterious	0	65	0.00000
CHR22	22	20090741	G	A	rs877782527	NO	missense	ENST00000238154.6:c.1216C>T	ENSP00000229178.6:p.Arg406Cys	R406C	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S28,7	benign	0.231	deleterious	0.05	15	0.00012
CHR22	22	20090742	G	T	rs877780171	NO	missense	ENST00000238154.6:c.1213C>A	ENSP00000229178.6:p.Arg405Ser	S405R	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S17,96	benign	0.016	tolerated	0.15	0	0.00018
CHR22	22	20090743	C	T	NA	NO	missense	ENST00000238154.6:c.1214A>G	ENSP00000229178.6:p.Asn405Ser	N405S	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S50,853	benign	0.001	tolerated	0.7	0	0.00000
CHR22	22	20090771	G	A	CM013842388	NO	missense	ENST00000238154.6:c.1186C>G	ENSP00000229178.6:p.Leu396Val	L396V	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S22,9	benign	0.211	tolerated	0.02	0	0.00000
CHR22	22	20090774	G	C	rs877780170	NO	missense	ENST00000238154.6:c.1183G>C	ENSP00000229178.6:p.Val395Leu	V395L	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S23,8	benign	0.138	deleterious	0.04	0	0.00015
CHR22	22	20090782	A	G	rs373073101,CM013511,COSM1535330	NO	missense	ENST00000238154.6:c.1171T>C	ENSP00000229178.6:p.Ser392Asp	S392D	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S24	probably_damaging	0.998	deleterious	0	35	0.00018
CHR22	22	20090787	G	T	CM004337182	YES	stop_gained	ENST00000238154.6:c.1170C>A	ENSP00000229178.6:p.Tyr390Ter	Y390*	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S39	NA	NA	NA	NA	NA	0.00000
CHR22	22	20090810	T	C	rs769439600	NO	missense	ENST00000238154.6:c.1147A>G	ENSP00000229178.6:p.Thr383Ala	T383A	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S26,6	probably_damaging	0.916	deleterious	0.03	25	0.00000
CHR22	22	20090816	TGAA	T	NA	YES	frameshift,feature_truncation	ENST00000238154.6:c.1139_1140delTC	ENSP00000229178.6:p.Leu380IleIleTer14	L380X	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S35	NA	NA	NA	NA	NA	0.00000
CHR22	22	20090856	AG	A	CS993415,rs55607708,KinMutBase,CHBKT	YES	frameshift,feature_truncation	ENST00000238154.6:c.1088delC	ENSP00000229178.6:p.Thr365ArgIleTer14	T365X	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S35	NA	NA	NA	NA	NA	0.00040
CHR22	22	20091865	A	G	rs877782840	NO	splice,intron	ENST00000238154.6:c.1096-47T>C	NA	NA	NA	13.68	NA	NA	NA	NA	0.00000
CHR22	22	20091871	A	G	NA	NO	intron	ENST00000238154.6:c.1096-105T>C	NA	NA	NA	15.76	NA	NA	NA	NA	0.00000
CHR22	22	20092870	C	T	rs200820484	NO	intron	ENST00000238154.6:c.1095+19G>A	NA	NA	NA	8.769	NA	NA	NA	NA	0.00000
CHR22	22	20092904	C	T	NA	NO	synonymous	ENST00000238154.6:c.1080C>A	ENSP00000229178.6:p.Ile360=	I360E	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S714,36	NA	NA	NA	NA	NA	0.00000
CHR22	22	20092908	T	C	rs766449049	NO	missense	ENST00000238154.6:c.1076A>G	ENSP00000229178.6:p.Glu359Gly	E359G	PROSITE_profiles:PS50011,PTHR12444:SF21,PTHR12444,3.10.510.10,Pfam_domain:PF00069:S22,8	benign	0.164	tolerated	0.07	0	0.00000
CHR22	22	20092914	G	A	rs765425451,CM118874	NO	missense	ENST00000238154.6:c.1070C>T	ENSP00000229178.6:p.Ser357Phe

CHR1C22	20130606	G	A	rs20106748	NO	missense	ENST00000132554.6:c.14C>T	ENSP00000129176.6:p.Ser5Leu	S5L	PTHR24344:SF21;PTHR24344	23.2	benign	0.003	deleterious	0.01	0	0.00015
CHR1C22	20130703	G	A	rs199708878	NO	missense	ENST00000132554.6:c.7C>T	ENSP00000129176.6:p.Arg3Trp	R3W	PTHR24344:SF21;PTHR24344	14.14	possibly_damaging	0.648	deleterious	0	25	0.00015
CHR1C22	20130713	G	A	rs1749381 48;CR068233	NO	splice_5_prime_UTR	ENST00000132554.6:c.-6C>T	NA	NA	NA	14.14	NA	NA	NA	NA	NA	0.00000
CHR1C22	20130724	G	C	rs56168060	NO	intron	ENST00000132554.6:c.-6-9C>G	NA	NA	NA	13.72	NA	NA	NA	NA	NA	0.00012
PAL B2 16	20164730	C	A	NA	NO	intron	ENST00000132554.6:c.930G>T	NA	NA	NA	9.745	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164789	G	C	NA	NO	missense	ENST00000132554.6:c.1353C>G	ENSP00000261154.4:p.His1184Gln	H1184Q	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	1.31	benign	0.018	tolerated	0.13	0	0.00000
PAL B2 16	20164792	G	C	CM070240,CM070239,rs11820990,PAL B2 c.3	YES	stop_gained	ENST00000132554.6:c.1354C>G	ENSP00000261154.4:p.Yr1183Ter	Y1183*	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	35	NA	NA	NA	NA	NA	0.00006
PAL B2 16	20164823	C	A	rs146635752	NO	missense	ENST00000132554.6:c.1383C>T	ENSP00000261154.4:p.Asn1177Ser	N1177S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	29.2	possibly_damaging	0.687	deleterious	0.01	0	0.00000
PAL B2 16	20164846	C	T	rs45439097	NO	synonymous	ENST00000132554.6:c.1495G>A	NA	S1165S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	18.52	NA	NA	NA	NA	NA	0.00216
PAL B2 16	20164850	A	G	rs72368500	NO	synonymous	ENST00000132554.6:c.1501A>G	NA	F1161F	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	0.153	NA	NA	NA	NA	NA	0.00003
PAL B2 16	20164908	C	T	NA	NO	missense	ENST00000132554.6:c.1613G>A	ENSP00000261154.4:p.Gly1145Ser	G1145S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	26.6	probably_damaging	1	tolerated	0.14	0	0.00000
PAL B2 16	20164913	A	C	CM111953,rs62652584	NO	missense	ENST00000132554.6:c.1618A>C	ENSP00000261154.4:p.Lys1144Ala	L1144H	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	24.8	probably_damaging	0.984	tolerated	0.09	0	0.00063
PAL B2 16	20164986	G	C	rs748338957	NO	missense	ENST00000132554.6:c.3355C>G	ENSP00000261154.4:p.Leu1119Val	L1119V	PTHR14662:SF2;PTHR14662	25.2	possibly_damaging	0.858	deleterious	0.01	25	0.00000
PAL B2 16	20164996	G	A	rs77098716	NO	splice_intron	ENST00000132554.6:c.3351-C	NA	NA	NA	5.812	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164997	G	T	NA	NO	splice_intron	ENST00000132554.6:c.3351-7C>A	NA	NA	NA	1.565	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165016	C	A	NA	NO	intron	ENST00000132554.6:c.3351-28G>A	NA	NA	NA	4.59	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165157	C	A	NA	NO	regulatory	ENST00000132554.6:c.3350+28G>T	NA	NA	NA	9.629	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165167	T	C	rs115726114	NO	regulatory	ENST00000132554.6:c.3350-11A>G	NA	NA	NA	5.929	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165182	T	C	NA	NO	splice_intron	ENST00000132554.6:c.3350-3A>G	NA	NA	NA	14.76	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165198	C	T	NA	NO	missense	ENST00000132554.6:c.3374C>A	ENSP00000261154.4:p.Gly1116Asp	G1116D	PTHR14662:SF2;PTHR14662	24.8	benign	0.24	deleterious	0.01	0	0.00000
PAL B2 16	20165225	C	T	rs148254555	NO	missense	ENST00000132554.6:c.3410G>A	ENSP00000261154.4:p.Gly1104Ser	G1104S	PTHR14662:SF2;PTHR14662	18.75	benign	0.129	tolerated	0.08	0	0.00000
PAL B2 16	20165228	C	G	rs201657283,CM118272	NO	missense	ENST00000132554.6:c.3413C>G	ENSP00000261154.4:p.Ser1103Cys	S1103L	PTHR14662:SF2;PTHR14662	15.35	benign	0.041	tolerated	0.08	0	0.00003
PAL B2 16	20165228	C	T	rs201657283,CM118272	NO	missense	ENST00000132554.6:c.3413C>A	ENSP00000261154.4:p.Val1101Met	V1101M	PTHR14662:SF2;PTHR14662	16.26	benign	0.043	tolerated	0.11	0	0.00003
PAL B2 16	20165235	A	C	rs45516180	NO	synonymous	ENST00000132554.6:c.3420T>G	NA	T1100T	PTHR14662:SF2;PTHR14662	0.059	NA	NA	NA	NA	NA	0.00571
PAL B2 16	20165238	C	T	rs45565738	NO	synonymous	ENST00000132554.6:c.3423G>A	NA	T1099T	PTHR14662:SF2;PTHR14662	18.68	NA	NA	NA	NA	NA	0.00036
PAL B2 16	20165239	G	A	rs142132127	NO	missense	ENST00000132554.6:c.3424C>G	ENSP00000261154.4:p.Thr1099Arg	T1099R	PTHR14662:SF2;PTHR14662	31	probably_damaging	0.971	deleterious	0	15	0.00012
PAL B2 16	20165253	C	T	rs74786102	NO	synonymous	ENST00000132554.6:c.3262G>A	NA	V1094V	PTHR14662:SF2;PTHR14662	13.77	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165279	G	A	rs587758527,CM092063,COSM31874	YES	stop_gained	ENST00000132554.6:c.3236C>T	ENSP00000261154.4:p.Arg1086Ter	R1086*	PTHR14662:SF2;PTHR14662	37	NA	NA	NA	NA	NA	0.00003
PAL B2 16	20165284	C	T	rs41578833	NO	missense	ENST00000132554.6:c.3232G>A	NA	S1084S	PTHR14662:SF2;PTHR14662	14.42	NA	NA	NA	NA	NA	0.00009
PAL B2 16	20165284	G	A	rs62652271	NO	missense	ENST00000132554.6:c.3231C>T	ENSP00000261154.4:p.Ser1084Leu	S1084L	PTHR14662:SF2;PTHR14662	13.18	benign	0.01	tolerated	0.37	0	0.00027
PAL B2 16	20165284	C	T	CM118271,rs147045425,COSM1637813	NO	missense	ENST00000132554.6:c.3230G>A	ENSP00000261154.4:p.Ser1083Asp	S1083D	PTHR14662:SF2;PTHR14662	23.1	benign	0.134	deleterious	0.02	15	0.00006
PAL B2 16	20165300	C	A	rs73088187 8	NO	missense	ENST00000132554.6:c.3215G>T	ENSP00000261154.4:p.Ala1078Ser	A1078S	PTHR14662:SF2;PTHR14662	22.7	benign	0.437	tolerated	0.18	0	0.00000
PAL B2 16	20165302	G	A	NA	NO	missense	ENST00000132554.6:c.3213C>T	ENSP00000261154.4:p.Cys1078Ser	C1078S	PTHR14662:SF2;PTHR14662	24	possibly_damaging	0.636	tolerated	0.4	0	0.00000
PAL B2 16	20165313	C	A	NA	NO	synonymous	ENST00000132554.6:c.3222G>T	NA	L1074L	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	16.85	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165315	C	G	NA	NO	missense	ENST00000132554.6:c.3220C>G	ENSP00000261154.4:p.Leu1074Val	L1074V	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	23.1	possibly_damaging	0.489	tolerated	0.06	0	0.00000
PAL B2 16	20165332	C	T	rs759587160	NO	missense,splice	ENST00000132554.6:c.3203G>A	ENSP00000261154.4:p.Gly1068Glu	G1068E	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	33	probably_damaging	0.994	deleterious	0	65	0.00003
PAL B2 16	20165341	T	A	rs367979106	NO	splice_intron	ENST00000132554.6:c.3202-8G>T	NA	NA	NA	3.79	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165341	C	T	rs367979106	NO	splice_intron	ENST00000132554.6:c.3202-8G>A	NA	NA	NA	3.715	NA	NA	NA	NA	NA	0.00003
PAL B2 16	20165342	G	A	rs75344247	NO	intron	ENST00000132554.6:c.3202-9C>T	NA	NA	NA	1.742	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165320	C	G	NA	NO	splice_intron	ENST00000132554.6:c.3201+5G>C	NA	NA	NA	13.28	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165480	A	T	rs148617733,CD070513,PAL B2 c.3116delA	YES	frameshift_indels,frame_truncation	ENST00000132554.6:c.3116delA	ENSP00000261154.4:p.Asn1039fsIleTer2	N1039X	PTHR14662:SF2;PTHR14662	35	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165855	G	T	rs145973006	NO	intron	ENST00000132554.6:c.3113+28C>A	NA	NA	NA	0.579	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165868	C	T	CM070242,rs1801771 32	YES	stop_gained,splice	ENST00000132554.6:c.3113C>A	ENSP00000261154.4:p.Ser1038Ter	W1038*	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	41	NA	NA	NA	NA	NA	0.00006
PAL B2 16	20165890	C	G	rs756906403	NO	missense	ENST00000132554.6:c.3106G>C	ENSP00000261154.4:p.Val1036Leu	V1036L	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	21.5	benign	0.092	tolerated	0.05	0	0.00006
PAL B2 16	20165938	T	C	NA	NO	missense	ENST00000132554.6:c.3088A>G	ENSP00000261154.4:p.Thr1030Ala	T1030A	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	25.1	probably_damaging	0.976	deleterious	0.01	55	0.00000
PAL B2 16	20165938	T	G	NA	NO	synonymous	ENST00000132554.6:c.3060A>G	ENSP00000261154.4:p.Thr1020Thr	Q1020Q	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	1.782	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165951	G	A	rs367948062	NO	missense	ENST00000132554.6:c.3044C>T	ENSP00000261154.4:p.Thr1015Ile	T1015I	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	26	probably_damaging	0.996	deleterious	0.01	0	0.00006
PAL B2 16	20165827	G	A	NA	NO	intron	ENST00000132554.6:c.2997-28C>T	NA	NA	NA	2.531	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165248	T	C	NA	NO	intron	ENST00000132554.6:c.2996+42A>G	NA	NA	NA	12.52	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165273	A	G	rs180177128	NO	synonymous	ENST00000132554.6:c.2996+17T>C	NA	NA	NA	18.26	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165295	C	T	rs455316;CM098533	NO	missense	ENST00000132554.6:c.2993G>A	ENSP00000261154.4:p.Gly998Glu	G998E	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	33	probably_damaging	0.992	deleterious	0	85	0.00438
PAL B2 16	20165308	G	A	rs61756146	NO	missense	ENST00000132554.6:c.2978C>T	ENSP00000261154.4:p.Thr993Met	T993M	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	24.7	possibly_damaging	0.743	tolerated	0.15	0	0.00000
PAL B2 16	20165315	C	A	rs755175665	NO	missense	ENST00000132554.6:c.2971G>T	ENSP00000261154.4:p.Val991F	V991F	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	23.1	possibly_damaging	0.822	deleterious	0.01	0	0.00003
PAL B2 16	20165324	G	A	CM070236,rs1820990,PAL B2 c.2962C>T	YES	stop_gained	ENST00000132554.6:c.2962C>T	ENSP00000261154.4:p.Gln988Ter	Q988*	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	36	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165339	T	A	rs587782211	NO	missense	ENST00000132554.6:c.2947A>T	ENSP00000261154.4:p.Thr983Ser	T983S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	13.27	benign	0.173	tolerated	0.42	0	0.00000
PAL B2 16	20165350	C	A	NA	NO	missense	ENST00000132554.6:c.2936G>T	ENSP00000261154.4:p.Ser979Ile	S979I	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	22.8	benign	0.197	deleterious	0.03	0	0.00000
PAL B2 16	20165364	CTT	C	rs180177126	YES	frameshift_feature_truncation	ENST00000132554.6:c.2920_2921delAA	ENSP00000261154.4:p.Lys974GlufsTer5	K974X	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	34	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165427	A	G	rs115726101	NO	synonymous	ENST00000132554.6:c.2859T>C	NA	D953D	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	2.72	NA	NA	NA	NA	NA	0.00003
PAL B2 16	20165435	A	G	rs149522812	NO	missense	ENST00000132554.6:c.2851T>C	ENSP00000261154.4:p.Ser951Pro	S951P	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	7.641	benign	0.045	tolerated	0.26	0	0.00018
PAL B2 16	20165446	A	G	rs454648580	NO	missense	ENST00000132554.6:c.2840T>C	ENSP00000261154.4:p.Leu947Ser	L947S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	25.5	probably_damaging	0.995	deleterious	0	65	0.00018
PAL B2 16	20165308	T	G	rs766701745	NO	non_coding_exon,utr	ENST00000132554.6:c.2834+22A>C	NA	NA	NA	7.802	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165311	C	T	NA	NO	non_coding_exon,utr	ENST00000132554.6:c.2834+18G>A	NA	NA	NA	4.659	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165317	G	T	rs515316577	NO	non_coding_exon,utr	ENST00000132554.6:c.2834+13C>A	NA	NA	NA	6.102	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165318	A	T	rs201817103	NO	missense	ENST00000132554.6:c.2816T>A	ENSP00000261154.4:p.Ile944Asn	I944N	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	31	probably_damaging	0.997	deleterious	0.01	0	0.00000
PAL B2 16	20165348	A	C	CM105609,rs45478192	NO	missense	ENST00000132554.6:c.2816T>G	ENSP00000261154.4:p.Leu939Trp	L939W	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	28	probably_damaging	1	deleterious	0	55	0.00018
PAL B2 16	20165368	A	G	rs748232940	NO	synonymous	ENST00000132554.6:c.2797C>T	NA	C933C	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	15.26	NA	NA	NA	NA	NA	0.00003
PAL B2 16	20165370	C	T	rs45624056,CM112100	NO	missense	ENST00000132554.6:c.2794G>A	ENSP00000261154.4:p.Val932Met	V932M	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	26.6	probably_damaging	0.995	tolerated	0.1	0	0.01617
PAL B2 16	20165412	G	A	CM118268	NO	missense	ENST00000132554.6:c.2723C>T	ENSP00000261154.4:p.Pro910Ser	P910S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	27.9	probably_damaging	1	deleterious	0.04	0	0.00000
PAL B2 16	20165727	G	A	NA	NO	intron	ENST00000132554.6:c.2748+38C>T	NA	NA	NA	6.547	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165741	C	T	rs73088187 3	NO	intron	ENST00000132554.6:c.2748+16G>A	NA	NA	NA	4.241	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165742	G	A	rs740756693	NO	intron	ENST00000132554.6:c.2748+15C>T	NA	NA	NA	1.334	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165744	G	C	rs771135581	NO	intron	ENST00000132554.6:c.2748+13C>G	NA	NA	NA	1.231	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165752	C	T	NA	NO	splice_intron	ENST00000132554.6:c.2748+5G>A	NA	NA	NA	11.85	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165760	G	A	rs115759782	NO	synonymous	ENST00000132554.6:c.2742C>T	ENSP00000261154.4:p.Ser914Ser	F914F	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	9.451	NA	NA	NA	NA	NA	0.00006
PAL B2 16	20165781	T	A	NA	NO	missense	ENST00000132554.6:c.2724A>T	ENSP00000261154.4:p.Lys908Asn	K908N	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	23.2	probably_damaging	0.998	tolerated	0.08	0	0.00000
PAL B2 16	20165787	T	C	CM111813,rs180177122	YES	stop_gained	ENST00000132554.6:c.2718G>A	ENSP00000261154.4:p.Trp906Ter	W906*	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	39	NA	NA	NA	NA	NA	0.00006
PAL B2 16	20165831	C	T	rs45476495	NO	missense	ENST00000132554.6:c.2674G>A	ENSP00000261154.4:p.Glu892Lys	E892K	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	33	probably_damaging	0.999	deleterious	0.05	0	0.00024
PAL B2 16	20165831	C	A	rs45476495	YES	stop_gained	ENST00000132554.6:c.2674G>T	ENSP00000261154.4:p.Glu892Ter	E892*	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	42	NA	NA	NA	NA	NA	0.00006
PAL B2 16	20165832	G	A	rs78179744	NO	synonymous	ENST00000132554.6:c.2673C>T	NA	C891C	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	9.786	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165843	T	C	NA	NO	missense	ENST00000132554.6:c.2662A>G	ENSP00000261154.4:p.Ile888Val	I888V	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	8.969	benign	0.034	tolerated	0.55	0	0.00000
PAL B2 16	20165845	A	C	rs45559935	NO	missense	ENST00000132554.6:c.2660T>G	ENSP00000261154.4:p.Ile887Ser	I887S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	29.9	probably_damaging	1	deleterious	0	15	0.00000
PAL B2 16	20165860	C	T	rs45460806	NO	missense	ENST00000132554.6:c.2645G>A	ENSP00000261154.4:p.Cys882Tyr	C882Y	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	0.256	benign	0.002	tolerated	0	0	0.00000
PAL B2 16	20165864	C	T	rs566315705	NO	missense	ENST00000132554.6:c.2641G>A	ENSP00000261154.4:p.Glu881Lys	G881S	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	26.6	probably_damaging	0.949	tolerated	0.11	0	0.00003
PAL B2 16	20165886	A	C	rs587782387	NO	missense	ENST00000132554.6:c.2619T>G	ENSP00000261154.4:p.Ser873Arg	S873R	PTHR14662:SF2;PTHR14662:2.130.10.10;Superfamily_domains:SSF50978	26.1	probably_damaging	0.998	deleterious	0.01	0	0.00000
PAL B2 16	20165898	G	A	rs73088187 0	NO	synonymous	ENST00000132554.6:c.2595T>C	ENSP00000261154.4:p.Val865Ala	V865A	PTHR14662:SF2;PTHR14662	27.3	probably_damaging	0.981	deleterious	0.02	15	0.00003
PAL B2 16	20165898	G	A	rs767633741,rs45542234	NO	synonymous	ENST00000132554.6:c.2607C>T	NA	S869S	PTHR14662:SF2;PTHR14662	14.93	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165898	G	C	rs767633741,rs45542234	NO	synonymous	ENST00000132554.6:c.2607C>G	NA	S869S	PTHR14662:SF2;PTHR14662	11.41	NA	NA	NA	NA	NA	0.00015
PAL B2 16	20165899	G	C	rs779279139	NO	missense	ENST00000132554.6:c.2606C>G	ENSP00000261154.4:p.Ser868Cys	S869C	PTHR14662:SF2;PTHR14662	27.7	probably_damaging	0.999	deleterious	0.02	15	0.00003
PAL B2 16	20165775	G	A	CM105610,rs45568339	NO	missense	ENST00000132554.6:c.2590C>T	ENSP00000261154.4:p.Pro864Ser	P864S	PTHR14662:SF2;PTHR14662	18.44	benign	0.348	tolerated	0.25	0	0.00006
PAL B2 16	20165754	T	C	rs76091408	NO	intron	ENST00000132554.6:c.2587-36A>G	NA	NA	NA	4.187	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165758	G	C	rs180177119	NO	intron	ENST00000132554.6:c.2587-38C>G	NA	NA	NA	1.441	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165758	G	C	NA	NO	intron	ENST00000132554.6:c.2587-40C>G	NA	NA	NA	1.667	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20165777	A	C	rs73088248	NO	intron	ENST00000132554.6:c.2587-595T>G	NA	NA	NA	4.083	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164867	G	A	rs249954	NO	intron	ENST00000132554.6:c.2586+38C>T	NA	NA	NA	5.159	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164868	T	C	rs78022695	NO	intron	ENST00000132554.6:c.2586+41A>G	NA	NA	NA	4.922	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164515	T	C	rs73322719	NO	intron	ENST00000132554.6:c.2586+188A>G	NA	NA	NA	11.84	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164519	T	C	rs144455175	NO	missense	ENST00000132554.6:c.2561A>G	ENSP00000261154.4:p.Asn854Ser	N854S	PTHR14662:SF2;PTHR14662	0.003	benign	0.023	tolerated	0.4	0	0.00003
PAL B2 16	20164589	GT	G	rs368990417,CD070510,rs180177116,PAL B2 c.	YES	frameshift_feature_truncation	ENST00000132554.6:c.2521delA	ENSP00000261154.4:p.Thr841GlufsTer10	T841X	PTHR14662:SF2;PTHR14662	22.7	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164963	G	C	rs75065768	NO	missense,splice	ENST00000132554.6:c.2485G>C	ENSP00000261154.4:p.Gln828Glu	Q828E	PTHR14662:SF2;PTHR14662	23.1	probably_damaging	0.998	tolerated	0.12	0	0.00000
PAL B2 16	20164967	G	A	NA	NO	synonymous	ENST00000132554.6:c.2598C>T	ENSP00000261154.4:p.Val866Val	V866V	PTHR14662:SF2;PTHR14662	6.117	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164969	C	T	CM084471001	NO	missense	ENST00000132554.6:c.2596G>A	ENSP00000261154.4:p.Val865Ile	V865I	PTHR14662:SF2;PTHR14662	0.001	benign	0.002	tolerated	0.72	0	0.00012
PAL B2 16	20164972	A	T	COSM3307838	NO	missense	ENST00000132554.6:c.2593T>A	ENSP00000261154.4:p.Ser865Thr	S865T	PTHR14662:SF2;PTHR14662	7.512	benign	0.297	tolerated	0.28	0	0.00000
PAL B2 16	20164997	G	A	NA	NO	synonymous	ENST00000132554.6:c.2478C>T	NA	N826N	PTHR14662:SF2;PTHR14662	1.473	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164022	A	G	NA	NO	missense	ENST00000132554.6:c.2453T>C	ENSP00000261154.4:p.Phe818Ser	F818S	PTHR14662:SF2;PTHR14662	0.594	benign	0.023	tolerated	0.4	0	0.00009
PAL B2 16	20164033	C	T	rs140776736	NO	synonymous	ENST00000132554.6:c.2442G>A	NA	E814E	PTHR14662:SF2;PTHR14662	10.09	NA	NA	NA	NA	NA	0.00012
PAL B2 16	20164037	A	G	rs763193051	NO	missense	ENST00000132554.6:c.2435T>C	ENSP00000261154.4:p.Ile813Thr	I813T	PTHR14662:SF2;PTHR14662	0.001	benign	0.002	tolerated	1	0	0.00006
PAL B2 16	20164058	G	A	rs45464991	NO	missense	ENST00000132554.6:c.2417C>T	ENSP00000261154.4:p.Pro806Leu	P806L	PTHR14662:SF2;PTHR14662	4.978	benign	0.023	tolerated	0.41	0	0.00006
PAL B2 16	20164089	C	A	CM070238,rs1801771 12	YES	stop_gained	ENST00000132554.6:c.2386G>T	ENSP00000261154.4:p.Gly796Ter	G796*	PTHR14662:SF2;PTHR14662	34	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164096	G	A	rs737826805	NO	synonymous	ENST00000132554.6:c.2379C>T	ENSP00000261154.4:p.Ser793Ser	G793G	PTHR14662:SF2;PTHR14662	3.292	NA	NA	NA	NA	NA	0.00018
PAL B2 16	20164146	C	T	rs148026749,CM112191154	NO	missense	ENST00000132554.6:c.2329G>A	ENSP00000261154.4:p.Asp777Asn	D777N	PTHR14662:SF2;PTHR14662	0.002	benign	0.006	tolerated	0.84	0	0.00000
PAL B2 16	20164147	G	A	rs45580997,CM096881 1	NO	synonymous	ENST00000132554.6:c.2328C>T	NA	F776F	PTHR14662:SF2;PTHR14662	9.552	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164149	AT	NA	NA	YES	frameshift_feature_elongation	ENST00000132554.6:c.2325dupA	ENSP00000261154.4:p.Phe776IleIleTer26	-775-776X	PTHR14662:SF2;PTHR14662	23.1	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164186	C	G	rs73347248	NO	missense	ENST00000132554.6:c.2280G>C	ENSP00000261154.4:p.Leu763Phe	L763F	PTHR14662:SF2;PTHR14662	17.95	possibly_damaging	0.897	tolerated	0.37	0	0.00000
PAL B2 16	20164218	G	A	rs180177116,CM070237,PAL B2 c. 2253C>T	YES	stop_gained	ENST00000132554.6:c.2253C>T	ENSP00000261154.4:p.Ser751Ser	R753*	PTHR14662:SF2;PTHR14662	35	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164219	T	C	rs147120218	NO	synonymous	ENST00000132554.6:c.2250A>G	NA	G752G	PTHR14662:SF2;PTHR14662	0.108	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164231	T	C	rs758046627	NO	synonymous	ENST00000132554.6:c.2244A>G	NA	T748T	PTHR14662:SF2;PTHR14662	0.132	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164275	T	A	rs45543843	NO	missense	ENST00000132554.6:c.2200A>T	ENSP00000261154.4:p.Thr734Ser	T734S	PTHR14662:SF2;PTHR14662	28.1	probably_damaging	1	deleterious	0	0	0.00000
PAL B2 16	20164388	T	A	NA	NO	missense	ENST00000132554.6:c.2047G>A	ENSP00000261154.4:p.Val683Ile	M723L	PTHR14662:SF2;PTHR14662	0.002	benign	0.001	tolerated	0.37	0	0.00000
PAL B2 16	20164314	T	TC	NA	YES	frameshift_feature_elongation	ENST00000132554.6:c.2160_2161insG	ENSP00000261154.4:p.Thr721AspfsTer24	-720-723X	PTHR14662:SF2;PTHR14662	23.3	NA	NA	NA	NA	NA	0.00000
PAL B2 16	20164340	G	A	rs414558731	NO	missense	ENST00000132554.6:c.2126G>A	ENSP00000261154.4:p.Ala712Val	A712V	PTHR14662:SF2;PTHR14662	0.353	benign	0.009	tolerated	0.17	0	0.00075
PAL B2 16	20164345	C	T	rs749409060,COSM40059424	NO	synonymous	ENST00000132554.6:c.2120G>A	NA	T710T	PTHR14662:SF2;PTHR14662	8.302	NA	NA	NA	NA	NA	0.00006
PAL B2 16	20164351	T	A	NA	NO	missense	ENST00000132554.6:c.2114A>T	ENSP00000261154.4:p.Leu705Phe	L706F	PTHR14662:SF2;PTHR14662	0.158	benign	0.077	tolerated	0.19	0	0.00000
PAL B2 16	20164369	T	C	rs73088186	NO	synonymous	ENST00000132554.6:c.2100A>G	ENSP00000261154.4:p.Ile702Met	I702M	PTHR14662:SF2;PTHR14662	0.003	benign	0.061	tolerated	0.3	0	0.00003
PAL B2 16	20164372	G	A	rs755387700	NO	synonymous	ENST00000132554.6:c.2103C>T	NA	S701S	PTHR14662:SF2;PTHR14662	7.164	NA	NA	NA	NA	NA	0.00003
PAL B2 16	20164375	T	A	rs75714584	NO	synonymous	ENST00000132554.6:c.2100A>T	NA	S700S	PTHR14662:SF2;PTHR14662	9.667	NA	NA	NA	NA	NA	0

PAL B2 16	23649405	G	C	rs15318635	NO	missense	ENST00000261584.4:c.94C>G	ENSP00000261584:p.Leu32Val	L32V	Coiled-coils_(Ncoils)|Coil,PTHR14662:SF2,PTHR14662	24.2	probably_damaging	0.998	deleterious	0	0	0.00003
PAL B2 16	23649415	G	A	rs761533286	NO	synonymous	ENST00000261584.4:c.84C>T	NA	Y28Y	Coiled-coils_(Ncoils)|Coil,PTHR14662:SF2,PTHR14662	4.639	NA	NA	NA	NA	0.00006
PAL B2 16	23649428	A	G	NA	NO	missense	ENST00000261584.4:c.71T>C	ENSP00000261584:p.Leu24Ser	L24S	Low_complexity_(Seg)|seg,Coiled-coils_(Ncoils)|Coil,PTHR14662:SF2,PTHR14662	27.9	probably_damaging	1	deleterious	0	65	0.00000
PAL B2 16	23649435	C	A	NA	NO	missense	ENST00000261584.4:c.64G>T	ENSP00000261584:p.Ala22Ser	A22S	Low_complexity_(Seg)|seg,Coiled-coils_(Ncoils)|Coil,PTHR14662:SF2,PTHR14662	25.6	probably_damaging	1	tolerated	0.06	0	0.00000
PAL B2 16	23649446	T	C	rs138786658	NO	missense	ENST00000261584.4:c.53A>G	ENSP00000261584:p.Lys18Arg	K18R	Low_complexity_(Seg)|seg,Coiled-coils_(Ncoils)|Coil,PTHR14662:SF2,PTHR14662	26.2	probably_damaging	1	deleterious	0	0	0.00012
PAL B2 16	23652332	C	T	NA	NO	regulatory	ENST00000261584.4:c.48+79G>A	NA	NA	NA	6.673	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652364	GC	G	NA	NO	intron	ENST00000261584.4:c.48+66delG	NA	NA	NA	8.117	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652369	G	A	NA	NO	regulatory	ENST00000261584.4:c.48+42C>T	NA	NA	NA	6.356	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652388	C	A	rs778858724	NO	regulatory	ENST00000261584.4:c.48+43G>T	NA	NA	NA	9.567	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652391	G	A	rs747578989	NO	regulatory	ENST00000261584.4:c.48C>T	NA	NA	NA	12	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652392	G	T	rs768839390	NO	regulatory	ENST00000261584.4:c.48+39C>A	NA	NA	NA	10.39	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652404	C	A	rs515726119	NO	regulatory	ENST00000261584.4:c.48+27G>T	NA	NA	NA	17.6	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652405	G	A	rs748775669	NO	regulatory	ENST00000261584.4:c.48+26C>T	NA	NA	NA	13.71	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652409	G	T	rs748775669	NO	regulatory	ENST00000261584.4:c.48+22C>A	NA	NA	NA	12.87	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652430	C	T	rs770451617	NO	regulatory	ENST00000261584.4:c.48+21G>A	NA	NA	NA	20.6	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652442	C	T	rs771287655	NO	missense	ENST00000261584.4:c.37G>A	ENSP00000261584:p.Glu13Lys	E13K	Low_complexity_(Seg)|seg,PTHR14662:SF2,PTHR14662	32	probably_damaging	1	tolerated	0.07	0	0.00003
PAL B2 16	23652464	G	A	COSMM158988	NO	synonymous	ENST00000261584.4:c.15C>T	NA	P5P	PTHR14662:SF2,PTHR14662	17.11	NA	NA	NA	NA	NA	0.00000
PAL B2 16	23652468	G	A	rs377386577,CM118266	NO	missense	ENST00000261584.4:c.13C>T	ENSP00000261584:p.Pro5Ser	P5S	PTHR14662:SF2,PTHR14662	8.381	benign	0.046	tolerated	0.55	0	0.00010
PAL B2 16	23652467	A	G	rs867706422	NO	synonymous	ENST00000261584.4:c.12T>C	NA	P4P	PTHR14662:SF2,PTHR14662	11.11	NA	NA	NA	NA	NA	0.00029
PAL B2 16	23652468	G	A	rs45619737	NO	missense	ENST00000261584.4:c.11C>T	ENSP00000261584:p.Pro4Leu	P4L	PTHR14662:SF2,PTHR14662	22.9	possibly_damaging	0.589	tolerated	0.10	0	0.00026
PAL B2 16	23652478	T	C	NA	NO	initiator_codon	ENST00000261584.4:c.1A>G	ENSP00000261584:p.Met1?	M1V	NA	22.3	benign	0.129	deleterious	0	NA	0.00000
XIRC17	23245728	A	G	rs3218539	NO	3_prime_UTR	ENST00000159321:1:c.7T>C	NA	NA	NA	12.68	NA	NA	NA	NA	NA	0.00000
XIRC17	23245759	TAAAA	T	NA	YES	frameshift_feature_truncation	ENST00000159321:1:c.807_810delTTTT	ENSP00000155271:1:p.Phe269LeufsTer27	F269-270X	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	35	NA	NA	NA	NA	NA	0.00000
XIRC17	23245779	T	C	rs58778812	NO	missense	ENST00000159321:1:c.811A>G	ENSP00000155271:1:p.Ile271Val	I271V	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	2.043	benign	0.003	tolerated	0.64	0	0.00003
XIRC17	23245762	A	C	rs145085742	NO	missense	ENST00000159321:1:c.808T>G	ENSP00000155271:1:p.Phe270Val	F270V	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	24.8	possibly_damaging	0.728	deleterious	0	45	0.00000
XIRC17	23245766	AT	A	rs776436748	YES	frameshift_feature_truncation	ENST00000159321:1:c.803delA	ENSP00000155271:1:p.His268LeufsTer29	H268X	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	28.3	NA	NA	NA	NA	NA	0.00000
XIRC17	23245797	C	T	rs149186933,COSMM206351	NO	missense	ENST00000159321:1:c.773G>A	ENSP00000155271:1:p.Arg258His	R258H	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	11.03	benign	0.002	tolerated	0.57	0	0.00003
XIRC17	23245798	G	A	rs759300252	NO	missense	ENST00000159321:1:c.772C>T	ENSP00000155271:1:p.Arg258Cys	R258C	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	18.26	benign	0.001	tolerated	0.21	0	0.00000
XIRC17	23245826	T	G	rs756589771	NO	missense	ENST00000159321:1:c.744A>C	ENSP00000155271:1:p.Glu248His	Q248H	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	0.218	benign	0.080	tolerated	0.13	0	0.00009
XIRC17	23245840	G	A	NA	YES	stop_gained	ENST00000159321:1:c.730C>T	ENSP00000155271:1:p.Gln244Ter	Q244*	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	35	NA	NA	NA	NA	NA	0.00000
XIRC17	23245858	T	A	NA	NO	missense	ENST00000159321:1:c.712A>T	ENSP00000155271:1:p.Arg238Trp	R238W	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	26.9	probably_damaging	1	deleterious	0	65	0.00000
XIRC17	23245901	G	A	rs374213093	NO	synonymous	ENST00000159321:1:c.669C>T	NA	V223V	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	0.087	NA	NA	NA	NA	NA	0.00003
XIRC17	23245903	A	G	rs878788130	NO	missense	ENST00000159321:1:c.667T>C	ENSP00000155271:1:p.Tyr223His	Y223H	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	17.79	probably_damaging	0.999	tolerated	0.05	0	0.00009
XIRC17	23245910	A	G	NA	NO	synonymous	ENST00000159321:1:c.660C>T	NA	D220D	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	0.03	NA	NA	NA	NA	NA	0.00000
XIRC17	23245917	TCA	T	rs74614129	YES	frameshift_feature_truncation	ENST00000159321:1:c.651_652delTG	ENSP00000155271:1:p.Cys217Ter	C217-218*	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	22.9	NA	NA	NA	NA	NA	0.00000
XIRC17	23245927	G	A	rs143153871	YES	stop_gained	ENST00000159321:1:c.643C>T	ENSP00000155271:1:p.Arg215Ter	R215*	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	34	NA	NA	NA	NA	NA	0.00015
XIRC17	23245930	G	A	NA	YES	stop_gained	ENST00000159321:1:c.640C>T	ENSP00000155271:1:p.Arg214Ter	R214*	PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF52540	34	NA	NA	NA	NA	NA	0.00000
XIRC17	23245945	G	T	rs780202744	NO	missense	ENST00000159321:1:c.625C>A	ENSP00000155271:1:p.Pro209Thr	P209T	Low_complexity_(Seg)|seg,PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF53.082	benign	0.186	tolerated	0.6	0	0.00003
XIRC17	23245950	T	C	rs61762969	NO	missense	ENST00000159321:1:c.620A>G	ENSP00000155271:1:p.Glu207Gly	E207G	Low_complexity_(Seg)|seg,PTHR22942:SF5,PTHR22942,3:40.50.300:Superfamily_domains:SSF53.437	benign	0.134	tolerated	0.21	0	0.00056
XIRC17	23245958	G	C	rs772419235	NO	missense	ENST00000159321:1:c.612C>G	ENSP00000155271:1:p.Ser204Arg	S204R	Low_complexity_(Seg)|seg,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:0.007	benign	0.049	tolerated	0.32	0	0.00003
XIRC17	23245965	G	A	NA	NO	missense	ENST00000159321:1:c.605C>T	ENSP00000155271:1:p.Ala202Val	A202V	Low_complexity_(Seg)|seg,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:2.561	benign	0.243	tolerated	0.54	0	0.00000
XIRC17	23245973	C	T	NA	NO	missense	ENST00000159321:1:c.597G>A	ENSP00000155271:1:p.Met199Ile	M199I	PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300:Superfamily_domains:SSF52540.23.6	possibly_damaging	0.676	tolerated	0.2	0	0.00000
XIRC17	23245989	G	A	rs775565256,COSMM148418	NO	missense	ENST00000159321:1:c.581C>T	ENSP00000155271:1:p.Thr194Met	T194M	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:26.6	probably_damaging	0.982	deleterious	0	15	0.00000
XIRC17	23245993	C	G	NA	NO	missense	ENST00000159321:1:c.577G>C	ENSP00000155271:1:p.Ala193Pro	A193P	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:25.6	probably_damaging	0.969	deleterious	0.01	25	0.00000
XIRC17	23246007	C	T	rs121835636,CM021368	NO	missense	ENST00000159321:1:c.563G>A	ENSP00000155271:1:p.Arg188His	R188H	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:15.95	benign	0.015	tolerated	0.28	0	0.13267
XIRC17	23246043	C	T	NA	NO	missense	ENST00000159321:1:c.527G>A	ENSP00000155271:1:p.Cys176Tyr	C176Y	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:26.3	probably_damaging	1	deleterious	0.01	0	0.00006
XIRC17	23246061	T	G	rs77814946	NO	missense	ENST00000159321:1:c.509A>C	ENSP00000155271:1:p.Glu170Ala	E170A	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:27.2	probably_damaging	1	deleterious	0.03	0	0.00006
XIRC17	23246080	C	A	NA	YES	stop_gained	ENST00000159321:1:c.490G>T	ENSP00000155271:1:p.Glu164Ter	E164*	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:38	NA	NA	NA	NA	NA	0.00000
XIRC17	23246088	T	C	NA	NO	missense	ENST00000159321:1:c.482A>G	ENSP00000155271:1:p.Asn161Ser	N161S	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:8.981	benign	0.203	tolerated	0.46	0	0.00000
XIRC17	23246093	G	A	rs202236401,COSMM1087678	NO	synonymous	ENST00000159321:1:c.477C>T	NA	R159R	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:7.052	NA	NA	NA	NA	NA	0.00021
XIRC17	23246094	C	T	rs536396806	NO	missense	ENST00000159321:1:c.476G>A	ENSP00000155271:1:p.Arg159His	R159H	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:32	probably_damaging	1	deleterious	0	25	0.00009
XIRC17	23246095	G	A	rs887788128	NO	missense	ENST00000159321:1:c.475C>T	ENSP00000155271:1:p.Arg159Cys	R159C	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:32	probably_damaging	1	deleterious	0	65	0.00000
XIRC17	23246114	T	G	rs758946940	NO	synonymous	ENST00000159321:1:c.456A>C	NA	S152S	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:5.151	NA	NA	NA	NA	NA	0.00000
XIRC17	23246120	G	C	rs771453207	NO	missense	ENST00000159321:1:c.450C>G	ENSP00000155271:1:p.Ser150Arg	S150R	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:25.3	probably_damaging	0.969	deleterious	0	65	0.00015
XIRC17	23246212	A	G	NA	NO	missense	ENST00000159321:1:c.354T>C	ENSP00000155271:1:p.Cys118Arg	C120R	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:23.6	probably_damaging	1	deleterious	0	0	0.00000
XIRC17	23246216	C	T	rs518977371	NO	synonymous	ENST00000159321:1:c.354G>A	NA	V118V	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:3.197	NA	NA	NA	NA	NA	0.00000
XIRC17	23246227	A	T	NA	NO	missense	ENST00000159321:1:c.343T>A	ENSP00000155271:1:p.Phe115Ile	F115I	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:0.033	benign	0.101	tolerated	0.06	0	0.00000
XIRC17	23246229	C	T	rs779741034	NO	missense	ENST00000159321:1:c.341G>A	ENSP00000155271:1:p.Arg114Lys	R114K	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:24.2	probably_damaging	0.999	deleterious	0	0	0.00003
XIRC17	23246270	T	C	rs146708851	NO	synonymous	ENST00000159321:1:c.300A>G	NA	L100L	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:0.016	NA	NA	NA	NA	NA	0.00000
XIRC17	23246284	G	T	rs778698188	NO	missense	ENST00000159321:1:c.286C>A	ENSP00000155271:1:p.Leu96Ile	L96I	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:24.5	probably_damaging	0.999	deleterious	0.01	0	0.00000
XIRC17	23246287	T	C	rs140214637	NO	missense	ENST00000159321:1:c.283A>G	ENSP00000155271:1:p.Ile95Val	I95V	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:0.001	benign	0.036	tolerated	0.18	0	0.00017
XIRC17	23246288	T	C	rs765998559	NO	synonymous	ENST00000159321:1:c.282A>G	NA	T94T	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:0.004	NA	NA	NA	NA	NA	0.00009
XIRC17	23246299	G	A	rs730882043	NO	missense	ENST00000159321:1:c.271C>T	ENSP00000155271:1:p.Arg91Trp	R91W	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:26.9	probably_damaging	1	deleterious	0	65	0.00003
XIRC17	23246322	G	GT	NA	YES	frameshift_feature_elongation	ENST00000159321:1:c.247dupA	ENSP00000155271:1:p.Thr83AsnfsTer6	T83NX	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:27.3	NA	NA	NA	NA	NA	0.00000
XIRC17	23246322	G	A	NA	NO	missense	ENST00000159321:1:c.248C>T	ENSP00000155271:1:p.Thr83Ile	T83I	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:22.6	probably_damaging	1	tolerated	0.25	15	0.00000
XIRC17	23246345	T	C	rs751657780	NO	synonymous	ENST00000159321:1:c.225A>G	NA	E75E	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:0.004	NA	NA	NA	NA	NA	0.00018
XIRC17	23246380	G	A	rs151110146	YES	stop_gained	ENST00000159321:1:c.190C>T	ENSP00000155271:1:p.Arg64Ter	R64*	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,Pfam_domain:PF08423,3:40.50.300,Sup:38	NA	NA	NA	NA	NA	0.00000
XIRC17	23246449	C	T	rs201836415	YES	splice_acceptor	ENST00000159321:1:c.122-1G>A	NA	NA	NA	24.8	NA	NA	NA	NA	NA	0.00000
XIRC17	23285788	T	C	rs777753452	NO	missense,splice	ENST00000159321:1:c.119A>G	ENSP00000155271:1:p.His40Arg	H40R	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942,3:40.50.300,Superfamily_domains:SSF121	possibly_damaging	0.529	tolerated	0.2	0	0.00000
XIRC17	23287792	C	T	rs730882040	NO	missense	ENST00000159321:1:c.115G>A	ENSP00000155271:1:p.Val39Met	V39M	PROSITE_profiles:PS50162,PTHR22942:SF5,PTHR22942	22.2	benign	0.33	tolerated	0.11	0	0.00003
XIRC17	23287842	C	T	NA	NO	missense	ENST00000159321:1:c.65G>A	ENSP00000155271:1:p.Ser22Asn	S22N	PROSITE_profiles:PS50162	22.7	benign	0.357	tolerated	0.17	0	0.00000
XIRC17	23287852	C	T	rs756828628	NO	missense	ENST00000159321:1:c.55G>A	ENSP00000155271:1:p.Glu19Lys	E19K	PROSITE_profiles:PS50162	27.3	probably_damaging	0.998	tolerated	0.06	0	0.00000
XIRC17	23287861	C	G	rs4987090	NO	missense	ENST00000159321:1:c.46G>C	ENSP00000155271:1:p.Ala16Pro	A16P	PROSITE_profiles:PS50162	28.9	probably_damaging	0.965	deleterious	0.01	0	0.00003
XIRC17	23287862	G	C	NA	NO	regulatory	ENST00000159321:1:c.39+64C>G	NA	NA	NA	5.93	NA	NA	NA	NA	NA	0.00000
XIRC17	23287863	G	A	rs764865989	NO	regulatory	ENST00000159321:1:c.39+43C>T	NA	NA	NA	7.786	NA	NA	NA	NA	NA	0.00000
XIRC17	23287880	T	C	NA	NO	regulatory	ENST00000159321:1:c.39+36A>G	NA	NA	NA	5.439	NA	NA	NA	NA	NA	0.00000
XIRC17	23287896	C	A	rs751787818	NO	regulatory	ENST00000159321:1:c.39+30G>T	NA	NA	NA	11.68	NA	NA	NA	NA	NA	0.00000
XIRC17	23287980	C	G	NA	NO	regulatory	ENST00000159321:1:c.39+26G>C	NA	NA	NA	8.588	NA	NA	NA	NA	NA	0.00000
XIRC17	23287980	C	T	NA	NO	regulatory	ENST00000159321:1:c.39+26G>A	NA	NA	NA	10.61	NA	NA	NA	NA	NA	0.00000
XIRC17	23287314	A	A	NA	NO	regulatory	ENST00000159321:1:c.39+12C>T	NA	NA	NA	15.6	NA	NA	NA	NA	NA	0.00000
XIRC17	23287121	C	G	NA	NO	splice_intron	ENST00000159321:1:c.39+5G>C	NA	NA	NA	20.5	NA	NA	NA	NA	NA	0.00000
XIRC17	23287125	C	T	rs764978131	YES	splice_donor	ENST00000159321:1:c.39+1G>A	NA	NA	NA	26.4	NA	NA	NA	NA	NA	0.00006
XIRC17	23273150	G	T	NA	NO	missense	ENST00000159321:1:c.15C>A	ENSP00000155271:1:p.Phe5Leu	F5L	NA	16.81	benign	0.004	tolerated	0.26	0	0.00000
XIRC17	23273165	C	T	rs768325997	NO	regulatory	ENST00000159321:1:c.-6G>A	NA	NA	NA	19.6	NA	NA	NA	NA	NA	0.00000
XIRC17	23273174	G	A	rs737867415	NO	regulatory	ENST00000159321:1:c.-18C>T	NA	NA	NA	13.39	NA	NA	NA	NA	NA	0.00000
XIRC17	23273178	C	T	rs750138805	NO	regulatory	ENST00000159321:1:c.-14G>A	NA	NA	NA	13.03	NA	NA	NA	NA	NA	0.00000
XIRC17	23273181	C	A	rs760456323	NO	regulatory	ENST00000159321:1:c.-17G>T	NA	NA	NA	10.36	NA	NA	NA	NA	NA	0.00000
XIRC17	23273181	C	T	rs760456323	NO	regulatory	ENST00000159321:1:c.-17G>A	NA	NA	NA	10.13	NA	NA	NA	NA	NA	0.00000
XIRC17	23273194	C	T	rs771060479	NO	regulatory	ENST00000159321:1:c.-30G>A	NA	NA	NA	10.58	NA	NA	NA	NA	NA	0.00000

ClinVar Significance	SEARCH Control Carrier Count	SEARCH Control Homozygous Carrier Count	SEARCH Control Carrier Frequency	SEARCH Case Carrier Count	SEARCH Case Homozygous Carrier Count	SEARCH Case Carrier Frequency
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	6	0	0.00046
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
uncertain_significance,benign,risk_factor	159	1	0.02897	274	4	0.02858
benign	43	0	0.00784	98	0	0.00749
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	2	0	0.00036	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	2	0	0.00036	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
uncertain_significance,likely_benign	1	0	0.00018	1	0	0.00008
likely_benign	2	0	0.00036	3	0	0.00023
pathogenic,benign	13	0	0.00237	39	0	0.00298
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	5	0	0.00091	25	0	0.00191
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
benign,likely_benign	4	0	0.00073	16	0	0.00122
NA	1	0	0.00018	0	0	0.00000
likely_benign	17	0	0.00310	62	0	0.00474
uncertain_significance	1	0	0.00018	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
benign,likely_benign	3	0	0.00055	15	0	0.00115
NA	1	0	0.00018	0	0	0.00000
NA	5	0	0.00091	10	0	0.00076
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	3	0	0.00055	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
benign	138	2	0.02515	332	3	0.02537
NA	1	0	0.00018	3	0	0.00023
uncertain_significance	1	0	0.00018	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	13	0	0.00237	39	0	0.00298
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
not_provided,likely_benign	11	0	0.00200	46	0	0.00351
uncertain_significance	1	0	0.00018	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	3	0	0.00023
NA	1	0	0.00018	8	0	0.00061
uncertain_significance,risk_factor	21	0	0.00383	85	0	0.00649
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign,pathogenic	0	0	0.00000	1	0	0.00008
uncertain_significance	4	0	0.00073	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
benign	1	0	0.00018	6	0	0.00046
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,likely_benign	29	0	0.00528	74	1	0.00565
NA	2	0	0.00036	4	0	0.00031
NA	1	0	0.00018	1	0	0.00008
benign	9	0	0.00164	32	0	0.00245
likely_benign	0	0	0.00000	6	0	0.00046
NA	0	0	0.00000	2	0	0.00015
likely_benign	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic,pathogenic	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	1	0	0.00018	0	0	0.00000
likely_pathogenic,pathogenic	0	0	0.00000	3	0	0.00023
NA	2	0	0.00036	1	0	0.00008
uncertain_significance	1	0	0.00018	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	1	0	0.00018	4	0	0.00031
uncertain_significance	1	0	0.00018	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
not_provided,benign	2	0	0.00036	10	0	0.00076
NA	0	0	0.00000	8	0	0.00061
uncertain_significance,not_provided	4	0	0.00073	2	0	0.00015
NA	2	0	0.00036	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
benign	1	0	0.00018	10	0	0.00076
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign,likely_benign	3	0	0.00055	6	0	0.00046
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	4	0	0.00073	9	0	0.00069
NA	0	0	0.00000	3	0	0.00023
uncertain_significance,not_provided,benign	19	0	0.00346	34	0	0.00260
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
likely_benign	12	0	0.00219	24	0	0.00183
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign	138	3	0.02510	345	6	0.02616
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
benign	1	0	0.00018	10	0	0.00076
likely_benign	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,pathogenic	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	4	0	0.00073	12	0	0.00092
uncertain_significance	1	0	0.00018	0	0	0.00000
benign,likely_benign	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,pathogenic	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	4	0	0.00073	12	0	0.00092
uncertain_significance	1	0	0.00018	0	0	0.00000
benign,likely_benign	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,pathogenic	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	2	0	0.00036	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
uncertain_significance	1	0	0.00018	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
benign	176	1	0.03207	396	8	0.03026
NA	2	0	0.00036	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
likely_benign	1	0	0.00018	2	0	0.00015
benign	1	0	0.00018	9	0	0.00069
NA	0	0	0.00000	1	0	0.00008

NA	0	0	0.00000	1	0	0.00000
benign	5	0	0.00091	25	0	0.00191
likely_benign	0	0	0.00000	2	0	0.00015
benign	1	0	0.00018	9	0	0.00069
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	3	0	0.00055	1	0	0.00008
likely_benign	10	0	0.00182	22	0	0.00168
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	84	0	0.01531	196	1	0.01498
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	5	0	0.00038
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	5	0	0.00091	4	0	0.00031
NA	3	0	0.00055	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	3	0	0.00023
uncertain_significance	9	0	0.00164	25	0	0.00191
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	5	0	0.00038
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	4	0	0.00031
NA	1	0	0.00018	0	0	0.00000
benign	317	2	0.05776	749	14	0.05723
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	1	0	0.00018	2	0	0.00015
NA	2	0	0.00036	2	0	0.00015
NA	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
benign	1507	119	0.27460	3482	252	0.26607
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
likely_benign	2	0	0.00036	5	0	0.00038
pathogenic	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	5	0	0.00038
uncertain_significance	0	0	0.00000	1	0	0.00008
likely_benign	2	0	0.00036	5	0	0.00038
likely_benign,pathogenic	2	0	0.00036	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
likely_benign	2	0	0.00036	1	0	0.00008
not_provided,benign	0	0	0.00000	5	0	0.00038
NA	16	0	0.00292	29	0	0.00222
NA	1	0	0.00018	0	0	0.00000
NA	2	0	0.00036	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	170	3	0.03098	438	12	0.03347
NA	2	0	0.00036	6	0	0.00046
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	1	0	0.00018	5	0	0.00038
likely_benign	4	0	0.00073	2	0	0.00015
NA	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
likely_benign	1	0	0.00018	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,not_provided	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	3	0	0.00023
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	27	0	0.00492	40	0	0.00306
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	98	0	0.01786	256	2	0.01956
NA	0	0	0.00000	4	0	0.00031
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
benign	0	0	0.00000	1	0	0.00008
uncertain_significance	3	0	0.00055	12	0	0.00092
uncertain_significance	5	0	0.00091	13	0	0.00099
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	2	0	0.00036	1	0	0.00008
NA	2	0	0.00036	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
not_provided,benign	5	0	0.00091	25	0	0.00191
uncertain_significance	3	0	0.00055	0	0	0.00000
likely_benign	9	0	0.00000	1	0	0.00008
benign	7	0	0.00128	8	0	0.00061
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign,likely_benign	205	6	0.03735	487	2	0.03721
uncertain_significance	1	0	0.00018	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	14	0	0.00255	18	0	0.00138
likely_benign	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance	2	0	0.00036	1	0	0.00008
uncertain_significance	1	0	0.00018	15	0	0.00115
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	2	0	0.00036	3	0	0.00023
uncertain_significance,not_provided,benign	4	0	0.00073	14	0	0.00107
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	2	0	0.00015
likely_benign	0	0	0.00000	2	0	0.00015
uncertain_significance	22	0	0.00401	36	0	0.00275
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	2	0	0.00015
benign	30	0	0.00547	76	0	0.00581
NA	1	0	0.00018	0	0	0.00000
likely_benign	2	0	0.00036	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
benign,likely_benign	555	18	0.09749	1280	33	0.09781
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	4	0	0.00073	2	0	0.00015
likely_benign	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	4	0	0.00031
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
benign	13	0	0.00237	39	0	0.00298
not_provided,likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance,not_provided	0	0	0.00000	1	0	0.00008
likely_benign	10	0	0.00182	15	0	0.00115
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
benign,likely_benign	1	0	0.00018	5	0	0.00038
NA	3	0	0.00055	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	13	0	0.00237	38	0	0.00290
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,not_provided	29	0	0.00528	53	0	0.00405
uncertain_significance	0	0	0.00000	7	0	0.00053
NA	0	0	0.00000	4	0	0.00031
NA	12	0	0.00219	36	0	0.00275
NA	0	0	0.00000	2	0	0.00015
NA	7	0	0.00128	31	0	0.00237
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,likely_pathogenic	0	0	0.00000	4	0	0.00031
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023

NA	2	0	0.00036	3	0	0.00023
likely_pathogenic,pathogenic	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	2	0	0.00036	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
likely_benign	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
benign	55	1	0.01002	134	0	0.01024
benign	341	6	0.06214	843	6	0.06442
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
benign	1512	120	0.27551	3484	253	0.26622
not_provided,benign,likely_benign	45	0	0.00784	133	0	0.01016
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
likely_benign	84	0	0.01531	196	1	0.01498
uncertain_significance	5	0	0.00091	4	0	0.00031
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance,pathogenic	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	3	0	0.00023
NA	1	0	0.00018	2	0	0.00015
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
not_provided,likely_benign	22	0	0.00401	45	0	0.00344
benign	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	3	0	0.00023
benign	1	0	0.00018	9	0	0.00069
NA	1	0	0.00018	0	0	0.00000
NA	2	0	0.00036	2	0	0.00015
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
pathogenic	3	0	0.00055	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
uncertain_significance,not_provided	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	2	0	0.00036	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	2	0	0.00015
not_provided,benign	5	0	0.00091	17	1	0.00130
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	62	0	0.01130	140	1	0.01070
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
benign	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
not_provided,benign	0	0	0.00000	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
benign	1	0	0.00018	8	0	0.00061
uncertain_significance	2	0	0.00036	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	4	0	0.00031
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	2	0	0.00015
likely_benign	8	0	0.00146	10	0	0.00076
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	4	0	0.00073	6	0	0.00046
NA	1	0	0.00018	0	0	0.00000
uncertain_significance,likely_benign	2	0	0.00036	2	0	0.00015
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
not_provided,benign	1	0	0.00018	15	0	0.00115
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	2	0	0.00036	2	0	0.00015
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
not_provided,benign	1	0	0.00018	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
likely_benign	3	0	0.00055	6	0	0.00046
benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	3	0	0.00055	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic,risk_factor	0	0	0.00000	8	0	0.00061
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	18	0	0.00328	16	0	0.00122
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	4	0	0.00031
likely_benign	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	2	0	0.00015
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	6	0	0.00046
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
pathogenic	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign,likely_benign	36	0	0.00656	77	0	0.00588
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance,not_provided	9	0	0.00164	8	0	0.00061
benign	2	0	0.00036	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	6	0	0.00046
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	2	0	0.00036	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008

pathogenic	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
benign	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic	1	0	0.00018	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic,pathogenic	0	0	0.00000	6	0	0.00046
uncertain_significance,not_provided	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	3	0	0.00055	5	0	0.00038
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	13	0	0.00237	40	0	0.00306
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign	9	0	0.00164	11	0	0.00084
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	7	0	0.00053
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,not_provided	1	0	0.00018	7	0	0.00053
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	3	0	0.00023
benign	478	14	0.08710	1149	29	0.08780
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	115	0	0.02095	234	2	0.01788
NA	3	0	0.00055	2	0	0.00015
NA	4641	1777	0.80922	10594	4225	0.80951
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	4	0	0.00073	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	0	0	0.00000
NA	2	0	0.00036	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	4494	1830	0.81888	10705	4376	0.81799
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	4	0	0.00031
NA	1	0	0.00018	2	0	0.00015
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic,pathogenic	1	0	0.00018	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
likely_pathogenic,pathogenic	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	7	0	0.00128	7	0	0.00053
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	1	0	0.00008
benign	5	0	0.00091	34	0	0.00260
not_provided,benign	1	0	0.00018	9	0	0.00069
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
uncertain_significance	2	0	0.00036	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
benign	0	0	0.00000	7	0	0.00053
uncertain_significance	2	0	0.00036	3	0	0.00023
NA	0	0	0.00000	2	0	0.00015
NA	4752	2201	0.86589	11201	5147	0.85589
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
not_provided,benign	1	0	0.00018	4	0	0.00031
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	6	0	0.00109	9	0	0.00069
pathogenic,risk_factor	3	0	0.00055	2	0	0.00015
uncertain_significance	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
pathogenic	3	0	0.00055	15	0	0.00115
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	5	0	0.00038
uncertain_significance	1	0	0.00018	5	0	0.00038
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
pathogenic	23	0	0.00419	173	0	0.01322
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	5	0	0.00038
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
pathogenic	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	1	0	0.00018	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
likely_pathogenic	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	2	0	0.00036	3	0	0.00023
NA	1	0	0.00018	1	0	0.00008
NA	2	0	0.00036	3	0	0.00023
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance,not_provided,pathogenic	1	1	0.00018	5	3	0.00038
uncertain_significance	1	0	0.00018	0	0	0.00000
not_provided	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
pathogenic	0	0	0.00000	4	0	0.00031
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	1	0	0.00008
uncertain_significance	2	0	0.00036	11	0	0.00084
uncertain_significance,pathogenic	0	0	0.00000	1	0	0.00008
uncertain_significance,pathogenic	2	0	0.00036	6	0	0.00046
uncertain_significance	8	0	0.00146	24	0	0.00183
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
uncertain_significance	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
likely_benign	3	0	0.00055	3	0	0.00023
uncertain_significance	2	0	0.00036	0	0	0.00000
not_provided,likely_pathogenic,pathogenic,risk_factor	4	0	0.00073	20	0	0.00153
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
not_provided	33	0	0.00601	54	0	0.00413
NA	1	0	0.00018	0	0	0.00000
not_provided,pathogenic	1	0	0.00018	2	0	0.00015
not_provided,pathogenic	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	1	0	0.00008
likely_benign	1	0	0.00018	7	0	0.00053
pathogenic	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
likely_pathogenic	3	0	0.00055	10	0	0.00076
uncertain_significance	1	0	0.00018	4	0	0.00031
uncertain_significance,not_provided	1	0	0.00018	33	0	0.00252
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1635	159	0.29792	3943	363	0.30129
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	5	0	0.00038
pathogenic	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
not_provided,pathogenic	0	0	0.00000	3	0	0.00023
benign	260	3	0.04738	655	13	0.05005
NA	0	0	0.00000	1	0	0.00008
NA	3	0	0.00055	12	0	0.00092
NA	1	0	0.00018	1	0	0.00008
uncertain_significance	2	0	0.00036	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	0	0	0.00000
not_provided	1	0	0.00018	0	0	0.00000

uncertain_significance,not_provided	4	0	0.00073	8	0	0.00061
uncertain_significance	2	0	0.00036	2	0	0.00015
uncertain_significance	0	0	0.00000	4	0	0.00031
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
not_provided,pathogenic,risk_factor	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
likely_benign	13	0	0.00237	57	0	0.00436
likely_benign	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	1	0	0.00018	6	0	0.00046
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
benign	347	1	0.06323	803	11	0.06136
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	2	0	0.00015
pathogenic	0	0	0.00000	4	0	0.00031
likely_benign	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
benign	0	0	0.00000	2	0	0.00015
benign	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
pathogenic,risk_factor	1	0	0.00018	8	0	0.00061
NA	0	0	0.00000	1	0	0.00008
pathogenic,risk_factor	2	0	0.00036	19	0	0.00145
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	2	0	0.00036	0	0	0.00000
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
benign,likely_benign	7	0	0.00128	15	0	0.00115
not_provided,benign	270	0	0.04920	591	4	0.04516
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic,risk_factor	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
pathogenic	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance,not_provided	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	9	0	0.00164	2	0	0.00015
uncertain_significance	17	0	0.00310	47	0	0.00359
NA	1	0	0.00018	0	0	0.00000
not_provided,benign	65	0	0.01184	155	0	0.01184
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
pathogenic	1	0	0.00018	7	0	0.00053
uncertain_significance	3	0	0.00055	8	0	0.00061
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
benign,likely_benign	0	0	0.00000	3	0	0.00023
benign,likely_benign	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
not_provided,likely_benign	44	0	0.00802	114	0	0.00871
NA	1	0	0.00018	2	0	0.00015
likely_benign	13	0	0.00237	23	0	0.00176
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
benign	2271	303	0.41381	5473	755	0.41805
NA	1	0	0.00018	1	0	0.00008
benign	3	0	0.00055	8	0	0.00061
NA	1	0	0.00018	0	0	0.00000
uncertain_significance,pathogenic	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
likely_benign	3	0	0.00055	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
pathogenic	0	0	0.00000	3	0	0.00023
uncertain_significance	1	0	0.00018	5	0	0.00038
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	2	0	0.00015
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance	2	0	0.00036	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
likely_benign	1	0	0.00018	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	2	0	0.00036	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	6	0	0.00046
not_provided,benign	346	1	0.06305	803	11	0.06136
NA	0	0	0.00000	1	0	0.00008
uncertain_significance,not_provided	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	2	0	0.00036	0	0	0.00000
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	7	0	0.00128	7	0	0.00053
NA	2	0	0.00036	1	0	0.00008
benign	0	0	0.00000	8	0	0.00061
NA	0	0	0.00000	1	0	0.00008
benign	923	41	0.16819	2136	89	0.16322
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
likely_benign	1	0	0.00018	2	0	0.00015
benign	61	1	0.01112	154	1	0.01177
NA	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	7	0	0.00128	12	0	0.00092
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	1	0	0.00008
uncertain_significance	5	0	0.00091	4	0	0.00031
uncertain_significance	1	0	0.00018	0	0	0.00000
pathogenic	0	0	0.00000	2	0	0.00015
benign	14	0	0.00255	42	0	0.00321
uncertain_significance	1	0	0.00018	5	0	0.00038
likely_benign	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
uncertain_significance	0	0	0.00000	3	0	0.00023
NA	0	0	0.00000	2	0	0.00015
pathogenic	0	0	0.00000	2	0	0.00015
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00000
uncertain_significance,not_provided,benign	229	4	0.04173	544	4	0.04157
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
not_provided,benign	3	0	0.00055	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
likely_benign	0	0	0.00000	2	0	0.00015
uncertain_significance,not_provided	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
likely_benign	1	0	0.00018	2	0	0.00015
pathogenic	0	0	0.00000	2	0	0.00015
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
not_provided,likely_benign	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	2	0	0.00036	9	0	0.00069
benign	0	0	0.00000	8	0	0.00061
pathogenic	0	0	0.00000	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
pathogenic	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
pathogenic	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
uncertain_significance,likely_benign	0	0	0.00000	3	0	0.00023
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	6	0	0.00046
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
pathogenic	1	0	0.00018	3	0	0.00023
likely_benign	2	0	0.00036	2	0	0.00015
uncertain_significance,not_provided	2	0	0.00036	6	0	0.00046
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	1	0	0.00018	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008

uncertain_significance,not_provided	1	0	0.00018	6	0	0.00046
likely_benign	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
not_provided,benign	2	0	0.00036	2	0	0.00015
NA	1	0	0.00018	5	0	0.00038
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	6	0	0.00046
NA	1	0	0.00018	0	0	0.00000
likely_benign	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	4	0	0.00031
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
likely_benign	2	0	0.00036	12	0	0.00092
uncertain_significance	1	0	0.00018	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	3	0	0.00055	33	1	0.00252
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	2	0	0.00015
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
uncertain_significance	3	0	0.00055	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	951	42	0.17529	2039	108	0.15580
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
benign	0	0	0.00000	1	0	0.00008
uncertain_significance	1	0	0.00018	2	0	0.00015
uncertain_significance	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	2	0	0.00015
uncertain_significance	0	0	0.00000	8	0	0.00061
NA	0	0	0.00000	1	0	0.00008
benign	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	5	0	0.00091	15	0	0.00115
benign	2	0	0.00036	5	0	0.00038
uncertain_significance	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
uncertain_significance	2	0	0.00036	0	0	0.00000
uncertain_significance	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	2	0	0.00015
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	3	0	0.00023
NA	1	0	0.00018	3	0	0.00023
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	1	0	0.00008
NA	1	0	0.00018	1	0	0.00008
NA	1	0	0.00018	0	0	0.00000
NA	0	0	0.00000	1	0	0.00008
NA	0	0	0.00000	2	0	0.00015