Supplemental Table 4: 169 HSP variants genotyped on the MS replication chip

Potentially Pathogenic Spastic Paraplegia Variants									UCSF Discovery cohort				NARCOMS cohort				Australian cohort				Italian cohort				Frequency in all cohorts				Enrichment vs ExAC European Cohort			Public allele frequencies					In-silico predicted pathogenicity					
MS Replication Chip SNP	Position	Ref	Alt	Variant Type	GENE	Transcript Variant(s)	Protein Variant(s)	Reported in literature	48 PPMS	203 SPMS	930 RMS	100 controls	122 PPMS	222 SPMS	586 RMS	321 controls	57 PPMS	75 SPMS	452 RMS	410 controls	87 PPMS	87 SPMS	280 RMS	156 controls	314 PPMS	587 SPMS	2248 RMS	987 controls	MAF in 314 PPMS / ExAC European	MAF in 587 SPMS / ExAC European	MAF in 2835 RMS / ExAC European	ExAC European	ExAC All cohorts	1000 Genomes	NHLBI ESP European	AFC	AFC European	CADD Score	Conservation phyloP p-value	IVA ACMG Classification	SIFT Function Prediction	SIFT Score	PolyPhen-2 Function Prediction	Reference in Literature
exm1701974	1:114438613	G	A	missense	*AP4B1*	c.1261C>T c.1054C>T c.1558C>T c.1508T>C	p.R421C p.R520C p.R352C p.I503T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.005%	0.007%	0.000%	0.000%	0.006%	0.007%	34	3.61E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm85551	1:114438882	A	G	missense	*AP4B1*	c.1211T>C c.1004T>C c.685A>G	p.I404T p.I335T p.I397V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.002%	0.000%	0.023%	0.001%	0.002%	24.4	5.59E-06	Uncertain Significance	Tolerated	0.1	Possibly Damaging	
exm85573	1:114441349	T	C	missense	*AP4B1*	c.1189A>G c.892A>G c.92G>A	p.I298V p.I229V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.004%	0.120%	0.439%	0.000%	0.100%	0.006%	11.29	6.90E-03	Uncertain Significance	Tolerated	0.22	Benign	
exm85617	1:114444457	C	T	missense	*AP4B1*	c.114-1435G>A c.389G>A c.61T>A	p.R130Q p.R31Q	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.010%	0.000%	0.000%	0.009%	0.001%	23.3	9.80E-04	Uncertain Significance	Tolerated	0.19	Possibly Damaging	
exm85619	1:114444488	A	T	missense	*AP4B1*	c.358T>A c.114-14667>A	p.Y120N p.Y21N	-	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	4.04	0.011%	0.008%	0.020%	0.012%	0.008%	0.011%	28.6	2.45E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm184791	2:32312629	G	A	missense	*SPAST*	c.484G>A	p.V162I	Non-pathogenic	0	0	2	0	0	1	1	0	0	0	3	2	0	0	0	0	0.000%	0.170%	0.267%	0.203%	0.00	0.57	0.90	0.297%	0.188%	0.020%	0.140%	0.152%	0.255%	19.06	2.30E-03	Uncertain Significance	Tolerated	0.48	Benign	https://bmcneurol.biomedcentral.com/articles/10.1186/1471-2377-10-17
exm184803	2:32339736	C	A	missense	*SPAST*	c.616C>A c.712C>A	p.P238T p.P206T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.010%	0.007%	0.000%	0.012%	0.006%	0.010%	22.1	1.99E-03	Uncertain Significance	Tolerated	0.56	Benign	
exm184804	2:32339767	G	A	missense	*SPAST*	c.743G>A c.647G>A	p.R248H p.R216H	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.001%	0.000%	0.000%	0.001%	0.000%	26.1	3.69E-05	Uncertain Significance	Damaging	0.04	Probably Damaging	
exm184830	2:32370014	A	G	missense	*SPAST*	c.1625A>G	p.D542G	+	0	1	2	0	0	0	1	0	0	1	0	0	0	0	0	0	0.000%	0.341%	0.133%	0.000%	0.00	5.59	2.19	0.061%	0.043%	0.020%	0.047%	0.040%	0.053%	24.5	3.46E-03	Uncertain Significance	Damaging	0.03	Benign	
exm241542	2:171675181	C	A	missense	*GAD1*	c.80C>A	p.T27K	-	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.101%	0.00	0.00	0.00	0.001%	0.075%	0.180%	0.000%	0.091%	0.001%	23.7	4.59E-05	Uncertain Significance	Tolerated	0.75	Benign	
exm241551	2:171686104	C	T	missense	*GAD1*	c.265C>T	p.R89W	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	0.78	0.057%	0.040%	0.020%	0.035%	0.039%	0.052%	34	3.60E-04	Uncertain Significance	Damaging	0.02	Possibly Damaging	
exm241556	2:171687535	A	T	missense	*GAD1*	c.380A>T	p.Y127F	-	0	1	2	0	0	0	2	0	0	0	3	3	0	0	0	0	0.000%	0.170%	0.311%	0.304%	0.00	5.01	9.16	0.034%	0.024%	0.000%	0.035%	0.023%	0.038%	23.2	1.40E-05	Uncertain Significance	Tolerated	0.45	Benign	
exm241562	2:171687639	C	G	missense	*GAD1*	c.484C>G	p.P162A	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.016%	0.000%	0.000%	0.013%	0.000%	23.4	1.37E-06	Uncertain Significance	Tolerated	0.45	Possibly Damaging	
exm241571	2:171700598	A	C	missense	*GAD1*	c.682A>C	p.I228L	-	1	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0.318%	0.000%	0.000%	0.101%	8.61	0.00	0.00	0.037%	0.026%	0.040%	0.047%	0.026%	0.042%	11.77	3.79E-04	Uncertain Significance	Tolerated	0.71	Benign	
exm255315	2:198352722	T	C	missense	*HSPD1*	c.1429A>G	p.M477V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.002%	0.000%	0.000%	0.001%	0.003%	17.85	1.01E-05	Uncertain Significance	Damaging	0.05	Benign	
exm255344	2:198361866	C	T	missense	*HSPD1*	c.425G>A	p.R142K	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.046%	0.060%	0.000%	0.040%	0.000%	23.4	2.45E-06	Uncertain Significance	Tolerated	0.09	Benign	
exm255359	2:198363406	C	T	missense	*HSPD1*	c.167G>A	p.G56E	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.106%	0.105%	0.000%	0.000%	0.186%	0.228%	26.3	1.49E-06	Uncertain Significance	Damaging	0.03	Possibly Damaging	
exm255361	2:198363437	G	A	missense	*HSPD1*	c.136C>T	p.L46F	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.024%	0.000%	0.000%	0.018%	0.001%	28.7	1.39E-06	Uncertain Significance	Damaging	0.03	Possibly Damaging	
exm282649	2:241658469	G	A	missense	*KIF1A*	c.4967C>T c.5168C>T c.4865C>T c.4892C>T c.4919C>T c.4651G>A c.4927G>A	p.A1631V p.A1640V p.A1656V p.A1723V p.A1622V p.D1542N p.D1576N	-	0	0	0	0	0	0	0	0	0	0	2	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	0.00	0.00	3.42	0.026%	0.023%	0.000%	0.035%	0.021%	0.028%	24.8	4.24E-06	Uncertain Significance	Tolerated	0.06	Benign	
exm282667	2:241659285	C	T	missense	*KIF1A*	c.4624G>A c.4679G>A c.4726G>A c.4586G>A c.4613G>A	p.D1643N p.D1560N p.D1551N p.R1630Q p.R1529Q	-	2	4	2	1	1	1	1	0	0	0	0	0	0	0	1	1	0.955%	0.852%	0.178%	0.203%	21.23	18.93	3.95	0.045%	0.039%	0.040%	0.036%	0.041%	0.044%	21.9	7.59E-06	Uncertain Significance	Tolerated	0.32	Benign	
exm282671	2:241659323	C	T	missense	*KIF1A*	c.4889G>A c.4688G>A c.4640G>A c.4403C>T c.4328C>T	p.R1547Q p.R1563Q p.R1538Q p.A1443V p.A1452V	-	1	3	4	2	0	0	0	1	2	0	0	0	0	0	0	0	0.955%	0.511%	0.178%	0.304%	2.69	1.44	0.50	0.355%	0.232%	0.180%	0.072%	0.194%	0.310%	27.6	4.74E-06	Uncertain Significance	Tolerated	0.64	Benign	
exm282694	2:241661934	G	A	missense	*KIF1A*	c.4604C>T c.4355C>T c.4301C>T c.3670C>T	p.A1535V p.A1468V p.A1434V p.R1190C	-	1	6	13	2	6	6	5	5	2	0	18	6	0	0	0	0	2.866%	2.044%	1.601%	1.317%	6.22	4.43	3.47	0.461%	0.244%	0.080%	0.516%	0.345%	0.631%	16.56	6.00E-04	Uncertain Significance	Tolerated	0.11	Benign	
exm282740	2:241679505	G	A	missense	*KIF1A*	c.3568C>T c.3595C>T c.3871C>T c.3494T>C	p.R1224C p.R1199C p.R1291C p.I1064T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.009%	0.040%	0.040%	0.012%	0.029%	0.005%	34	5.73E-03	Uncertain Significance	Damaging	0.05	Possibly Damaging	
exm282764	2:241683449	A	G	missense	*KIF1A*	c.3218T>C c.3191T>C c.3293T>C c.2809G>A	p.I1073T p.I1098T p.I1165T p.V928M	-	0	0	2	0	0	1	2	0	0	0	0	0	0	0	0	2	0.000%	0.170%	0.178%	0.203%	0.00	2.54	2.66	0.067%	0.046%	0.040%	0.130%	0.046%	0.072%	23	2.05E-05	Uncertain Significance	Tolerated	0.06	Benign	
exm282783	2:241685573	C	T	missense	*KIF1A*	c.2884G>A c.2782G>A c.3085G>A c.2750A>C	p.V1029M p.V962M p.V937M p.H1018P	-	0	1	2	1	2	0	3	2	1	1	2	0	0	0	1	0	0.955%	0.341%	0.356%	0.304%	2.46	0.88	0.92	0.388%	0.221%	0.060%	0.264%	0.209%	0.339%	23	4.82E-03	Uncertain Significance	Tolerated	0.2	Benign	
exm282785	2:241686663	T	G	missense	*KIF1A*	c.2852A>C c.2777A>C c.3053A>C	p.H917P p.H951P p.H926P	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.021%	0.013%	0.000%	0.012%	0.009%	0.017%	21.5	1.59E-03	Uncertain Significance	Tolerated	0.27	Benign	
exm282858	2:241722018	C	A	stopgain	*KIF1A*	c.871G>T	p.G291*	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.000%	0.000%	0.000%	0.000%	0.000%	37	2.23E-05	Uncertain Significance	-	-	-	
exm360575	3:155571417	G	A	missense	*SLC33A1*	c.370C>T	p.L124F	-	0	0	0	0	1	0	2	0	0	0	2	0	0	0	0	0	0.318%	0.000%	0.178%	0.000%	8.61	0.00	4.81	0.037%	0.027%	0.020%	0.035%	0.027%	0.039%	24	5.16E-03	Uncertain Significance	Tolerated	0.09	Possibly Damaging	
exm360585	3:155571651	C	T	missense	*SLC33A1*	c.136G>A	p.E46K	-	0	0	4	0	0	0	1	0	0	1	1	2	1	1	0	1	0.318%	0.341%	0.267%	0.304%	1.07	1.15	0.90	0.297%	0.208%	0.040%	0.140%	0.167%	0.261%	22.9	4.33E-06	Uncertain Significance	Tolerated	0.42	Benign	
exm417771	4:108866306	T	C	missense	*CYP2U1*	c.671T>C	p.I224T	-	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	1	0.000%	0.000%	0.000%	0.203%	0.00	0.00	0.00	0.010%	0.006%	0.000%	0.023%	0.005%	0.009%	23.5	2.10E-05	Uncertain Significance	Damaging	0.01	Benign	
exm417789	4:108868537	G	A	missense	*CYP2U1*	c.1132G>A	p.V378I	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.035%	0.120%	0.000%	0.030%	0.000%	23.3	4.61E-05	Uncertain Significance	Tolerated	0.11	Benign	
exm417803	4:108870614	G	A	missense	*CYP2U1*	c.1397G>A	p.R466Q	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.015%	0.040%	0.000%	0.011%	0.000%	35	1.51E-06	Uncertain Significance	Damaging	0.02	Probably Damaging	
exm2150936	7:4821257	G	A	missense	*AP5Z1*	c.238G>A	p.E80K	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.007%	0.007%	0.040%	0.024%	0.008%	0.006%	23.2	5.30E-03	Uncertain Significance	Damaging	0.04	Possibly Damaging	
exm601637_ver3	7:4820915	A	G	missense	*AP5Z1*	c.151A>G	p.I51V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.002%	0.040%	0.000%	0.001%	0.002%	13.39	2.51E-03	Uncertain Significance	Tolerated	0.27	Benign	
exm601639	7:4820940	G	A	missense	*AP5Z1*	c.176G>A	p.R59Q	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.017%	0.020%	0.000%	0.012%	0.016%	0.014%	28.8	1.02E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm601666	7:4823008	G	A	missense	*AP5Z1*	c.428G>A	p.R143Q	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	11.12	0.004%	0.101%	0.280%	0.012%	0.090%	0.007%	25.8	9.73E-03	Uncertain Significance	Damaging	0	Possibly Damaging	
exm601687	7:4823883	C	T	missense	*AP5Z1*	c.671C>T	p.T224M	-	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	1.71	0.026%	0.035%	0.000%	0.036%	0.023%	0.023%	27.5	1.20E-05	Uncertain Significance	Damaging	0	Damaging	
exm601713	7:4824629	G	A	missense	*AP5Z1*	c.881G>A	p.R294Q	-	0	1	3	1	1	0	5	2	1	0	2	3	0	0	3	1	0.637%	0.170%	0.578%	0.709%	1.25	0.34	1.14	0.508%	0.334%	0.120%	0.317%	0.312%	0.493%	26.7	3.18E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm601724	7:4825069	G	A	missense	*AP5Z1*	c.955G>A	p.D319N	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.004%	0.007%	0.040%	0.012%	0.005%	0.003%	23	1.45E-04	Uncertain Significance	Tolerated	0.15	Benign	
exm601798	7:4828518	G	T	missense	*AP5Z1*	c.1643G>T	p.R548L	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.001%	0.000%	0.000%	0.001%	0.000%	29.9	1.31E-05	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm601799	7:4828541	G	A	missense	*AP5Z1*	c.1666G>A	p.V556M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.020%	0.040%	0.000%	0.016%	0.002%	25.2	3.90E-03	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm601829	7:4830220	G	A	missense	*AP5Z1*	c.1936G>A	p.V646M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.132%	0.180%	0.000%	0.110%	0.001%	27.2	2.89E-03	Uncertain Significance	Damaging	0	Possibly Damaging	
exm601833	7:4830329	C	T	missense	*AP5Z1*	c.1964C>T	p.S655L	-	0	1	1	0	0	0	2	1	0	0	0	0	0	2	0	0	0.000%	0.511%	0.133%	0.101%	0.00	4.56	1.19	0.112%	0.069%	0.000%	0.108%	0.058%	0.102%	26.2	1.47E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm601836	7:4830350	G	A	missense	*AP5Z1*	c.1985G>A	p.C662Y	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.004%	0.040%	0.000%	0.005%	0.000%	27.4	3.83E-06	Uncertain Significance	Damaging	0	Probably Damaging	
exm601839	7:4830379	G	A	missense	*AP5Z1*	c.2014G>A	p.E672K	-	0	0	1	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	0.00	0.00	8.09	0.011%	0.008%	0.080%	0.012%	0.007%	0.011%	35	3.83E-06	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm601842	7:4830425	C	G	missense	*AP5Z1*	c.2060C>G	p.S687C	-	0	0	1	0	0	0	0	0	0	0	1	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	0.00	0.00	6.84	0.013%	0.008%	0.000%	0.000%	0.008%	0.013%	24.3	2.15E-03	Uncertain Significance	Damaging	0.03	Possibly Damaging	
exm601846	7:4830460	A	C	missense	*AP5Z1*	c.2095A>C	p.T699P	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.000%	0.000%	0.000%	0.000%	0.000%	23.3	1.62E-03	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm601850	7:4830496	C	G	missense	*AP5Z1*	c.2131C>G	p.R711G	-	0	0	3	0	1	0	0	2	0	3	1	0	0	0	0	1	0.318%	0.511%	0.178%	0.304%	2.38	3.81	1.33	0.134%	0.085%	0.000%	0.108%	0.089%	0.134%	33	3.63E-04	Uncertain Significance	Damaging	0	Possibly Damaging	
exm601876	7:4830879	G	A	missense	*AP5Z1*	c.2287G>A	p.V763M	-	1	0	1	0	0	0	0	0	0	0	0	0	1	0	0	0	0.637%	0.000%	0.044%	0.000%	70.77	0.00	4.94	0.009%	0.010%	0.020%	0.035%	0.015%	0.010%	29.5	1.75E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm639592	7:99701087	C	T	missense	*AP4M1*	c.407C>T	p.T136M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.010%	0.020%	0.000%	0.007%	0.001%	25.1	1.91E-05	Uncertain Significance	Tolerated	0.13	Benign	

exm639596	7:99701268	C	T	missense	*AP4M1*	c.496C>T	p.P166S	-	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.101%	0.00	0.00	0.00	0.007%	0.004%	0.000%	0.012%	0.004%	0.006%	25.4	1.75E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm639605	7:99702501	C	T	missense	*AP4M1*	c.611C>T	p.S204F	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.002%	0.000%	0.000%	0.002%	0.003%	23.2	1.98E-03	Uncertain Significance	Tolerated	0.69	Possibly Damaging	
exm639647	7:99704060	G	C	missense	*AP4M1*	c.1060G>C	p.A354P	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.043%	0.026%	0.000%	0.012%	0.021%	0.036%	27.7	1.58E-05	Uncertain Significance	Tolerated	0.18	Probably Damaging	
exm639648	7:99704079	C	A	missense	*AP4M1*	c.1079C>A	p.A360D	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.019%	0.040%	0.000%	0.016%	0.001%	22.1	9.29E-03	Uncertain Significance	Tolerated	0.42	Benign	
exm639649	7:99704100	G	A	missense	*AP4M1*	c.1100G>A	p.R367Q	-	1	0	1	0	0	0	1	0	0	0	1	0	0	0	0	0	0.318%	0.000%	0.133%	0.000%	7.08	0.00	2.97	0.045%	0.032%	0.020%	0.058%	0.029%	0.045%	34	1.59E-05	Uncertain Significance	Tolerated	0.11	Possibly Damaging	
exm2167516	8:17137944	C	G	missense	*VPS37A*	c.961C>G c.886C>G	p.L321V p.L296V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.004%	0.020%	0.000%	0.012%	0.000%	26.4	1.32E-04	Uncertain Significance	Damaging	0.04	Benign	
exm2167520	8:17143910	C	T	missense	*VPS37A*	c.1168C>T c.1093C>T	p.H390Y p.H365Y	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.020%	0.080%	0.000%	0.024%	0.000%	25.5	1.49E-05	Uncertain Significance	Tolerated	0.22	Benign	
exm2246677	8:17142033	G	A	missense	*VPS37A*	c.1021G>A c.946G>A	p.A341T p.A316T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.002%	0.000%	0.000%	0.003%	0.000%	33	2.64E-06	Uncertain Significance	Damaging	0	Possibly Damaging	
exm684255	8:17133963	C	A	missense	*VPS37A*	c.700C>A c.625C>A	p.L234I p.L209I	-	1	0	1	0	1	1	1	0	0	0	0	0	0	0	0	0	0.637%	0.170%	0.089%	0.000%	9.65	2.58	1.35	0.066%	0.041%	0.000%	0.058%	0.036%	0.061%	34	2.31E-05	Uncertain Significance	Damaging	0.05	Possibly Damaging	
exm684273	8:17137912	C	T	missense	*VPS37A*	c.854C>T c.929C>T	p.T285I p.T310I	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.007%	0.000%	0.000%	0.006%	0.000%	21.5	1.41E-03	Uncertain Significance	Tolerated	0.14	Benign	
exm696249	8:38091917	G	T	missense	*DDHD2*	c.226G>T	p.G76C	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.060%	0.139%	0.319%	0.058%	0.120%	0.054%	23.5	2.43E-03	Uncertain Significance	Tolerated	0.08	Benign	
exm696278	8:38103310	C	T	missense	*DDHD2*	c.899C>T	p.T300I	-	0	0	2	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	0.00	0.00	4.68	0.019%	0.012%	0.000%	0.012%	0.013%	0.020%	23	5.72E-05	Uncertain Significance	Damaging	0.05	Benign	
exm696290	8:38103783	C	G	missense	*DDHD2*	c.1090C>G	p.Q364E	-	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0.318%	0.000%	0.000%	0.000%	63.69	0.00	0.00	0.005%	0.003%	0.000%	0.023%	0.004%	0.008%	25.8	6.71E-06	Uncertain Significance	Damaging	0	Probably Damaging	
exm696329	8:38111229	T	A	missense	*DDHD2*	c.2047T>A	p.C683S	-	0	0	3	0	2	0	5	1	0	2	4	0	0	0	1	0	0.637%	0.341%	0.578%	0.101%	1.34	0.72	1.21	0.476%	0.297%	0.100%	0.221%	0.255%	0.425%	28.7	3.87E-05	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm703395	8:65509264	G	A	missense	*CYP7B1*	c.1456C>T c.1234- 6863C>T	p.R486C	HSP5	0	0	1	1	0	1	0	0	0	2	2	0	0	0	0	0	0.000%	0.511%	0.133%	0.101%	0.00	4.69	1.22	0.109%	0.070%	0.060%	0.116%	0.070%	0.117%	29	4.35E-05	Likely Pathogenic	Damaging	0	Probably Damaging	https://www.ncbi.nlm.nih.gov/pubmed/19439420
exm749625	9:35738127	G	T	missense	*GBA2*	c.2220C>A	p.S740R	-	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0.000%	0.000%	0.000%	0.101%	0.00	0.00	0.00	0.068%	0.048%	0.040%	0.058%	0.058%	0.087%	24.4	7.03E-07	Uncertain Significance	Tolerated	0.34	Possibly Damaging	
exm749661	9:35739712	C	T	missense	*GBA2*	c.1495G>A	p.E499K	-	0	0	2	0	1	1	1	0	0	0	0	0	0	0	0	0	0.318%	0.170%	0.133%	0.000%	12.74	6.81	5.34	0.025%	0.015%	0.000%	0.035%	0.015%	0.025%	18.16	3.37E-04	Uncertain Significance	Tolerated	0.23	Benign	
exm749675	9:35740293	C	G	missense	*GBA2*	c.1196G>C	p.R399P	-	1	2	4	0	0	0	2	2	1	3	3	4	0	0	0	0	0.637%	0.852%	0.400%	0.608%	3.42	4.58	2.15	0.186%	0.131%	0.000%	0.186%	0.129%	0.190%	12.35	4.91E-03	Uncertain Significance	Activating	1	Benign	
exm749695	9:35741033	A	G	missense	*GBA2*	c.815T>C	p.V272A	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.032%	0.160%	0.000%	0.028%	0.000%	25.9	6.14E-06	Uncertain Significance	Tolerated	0.07	Probably Damaging	
exm749700	9:35741796	C	T	missense	*GBA2*	c.659G>A	p.G220D	-	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0.000%	0.000%	0.000%	0.101%	0.00	0.00	0.00	0.014%	0.009%	0.000%	0.000%	0.014%	0.024%	25.8	7.94E-07	Uncertain Significance	Tolerated	0.2	Probably Damaging	
exm847062	10:99498266	C	T	missense	*ZFYVE27*	c.32C>T c.- 27+1188C>T	p.P11L	-	0	1	0	0	0	0	0	0	0	0	0	0	1	0	1	0	0.318%	0.170%	0.044%	0.000%	22.75	12.17	3.18	0.014%	0.154%	0.619%	0.023%	0.138%	0.012%	23.5	3.30E-03	Uncertain Significance	Tolerated	0.26	Benign	
exm847076	10:99502891	C	T	missense	*ZFYVE27*	c.238C>T c.173- 1595C>T c.197+4460C >T c.-26- 1595C>T	p.L80F	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.048%	0.120%	0.012%	0.043%	0.000%	25.5	1.44E-03	Uncertain Significance	Tolerated	0.11	Possibly Damaging	
exm847085	10:99504641	C	T	missense	*ZFYVE27*	c.198- 4590C>T c.130C>T c.198- 3385C>T c.424C>T c.328C>T	p.R44C p.R142C p.R110C	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	8.90	0.005%	0.024%	0.020%	0.035%	0.019%	0.005%	33	4.72E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm847132	10:99519057	A	G	stoploss	*ZFYVE27*	c.1251A>G c.861A>G c.1236A>G c.1215A>G c.936A>G c.957A>G c.1119A>G	p.*287W p.*373W p.*417W p.*319W p.*405W p.*412W p.*312W	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.000%	0.000%	0.012%	0.002%	0.006%	19.87	2.38E-04	Uncertain Significance	-	-	-	
exm1016848	12:57962833	G	A	missense	*KIF5A*	c.802G>A	p.A268T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.002%	0.040%	0.012%	0.001%	0.001%	32	2.97E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1016858	12:57963802	G	C	missense	*KIF5A*	c.1150G>C	p.G384R	-	2	1	1	0	0	0	2	0	0	0	0	0	0	0	0	0	0.637%	0.170%	0.133%	0.000%	16.76	4.48	3.51	0.038%	0.039%	0.040%	0.070%	0.037%	0.039%	19.07	2.15E-03	Uncertain Significance	Tolerated	0.5	Benign	
exm1016904	12:57970617	G	A	missense	*KIF5A*	c.2272G>A	p.E758K	-	0	1	5	0	1	0	0	0	0	0	0	0	0	0	0	0	0.318%	0.170%	0.222%	0.000%	4.03	2.16	2.82	0.079%	0.055%	0.000%	0.116%	0.058%	0.090%	24	2.37E-04	Uncertain Significance	Tolerated	0.34	Benign	
exm1047775	12:123741403	G	A	missense	*C12orf65*	c.326G>A	p.R109Q	-	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.170%	0.000%	0.000%	NaN	NaN	Inf	0.000%	0.003%	0.000%	0.000%	0.002%	0.000%	35	1.47E-06	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm1047779	12:123741490	A	G	missense	*C12orf65*	c.413A>G	p.K138R	-	0	0	1	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	0.00	0.00	8.09	0.011%	0.045%	0.140%	0.023%	0.043%	0.014%	22.3	3.29E-05	Uncertain Significance	Tolerated	0.07	Probably Damaging	
exm1063430	13:36909469	G	A	missense	*SPG20*	c.499C>T	p.P167S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.027%	0.017%	0.020%	0.000%	0.013%	0.023%	24.4	9.10E-04	Uncertain Significance	Damaging	0.02	Probably Damaging	
exm2221617	13:36878734	G	A	missense	*SPG20*	c.1769C>T	p.A590V	-	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0.000%	0.170%	0.000%	0.000%	0.00	42.59	0.00	0.004%	0.036%	0.040%	0.000%	0.034%	0.003%	24.1	1.97E-04	Uncertain Significance	Damaging	0	Benign	
exm1095315	14:31535541	G	A	Splice Site	*AP4S1*	c.138+1G>A	-	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.005%	0.003%	0.000%	0.012%	0.002%	0.004%	24.7	5.46E-06	Uncertain Significance	-	-	-	
exm1095316	14:31539064	T	C	missense	*AP4S1*	c.154T>C	p.Y52H	-	1	0	4	0	0	0	0	0	0	0	0	0	0	0	0	0	0.318%	0.000%	0.178%	0.000%	2.90	0.00	1.62	0.110%	0.068%	0.000%	0.116%	0.061%	0.097%	21.9	4.15E-05	Uncertain Significance	Tolerated	0.5	Probably Damaging	
exm1100690	14:51054710	G	C	missense	*ATL1*	c.196G>C	p.E66Q	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.015%	0.009%	0.000%	0.012%	0.009%	0.015%	15.75	7.33E-07	Uncertain Significance	Tolerated	0.29	Benign	
exm1100693	14:51057698	A	G	missense	*ATL1*	c.322A>G	p.T108A	Hereditary Sensory Neuropathy	0	0	0	0	0	0	0	0	0	0	2	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	0.00	0.00	2.17	0.041%	0.025%	0.000%	0.000%	0.018%	0.033%	15.45	9.71E-03	Uncertain Significance	Tolerated	0.42	Benign	https://www.omim.org/entry/606439
exm1100725	14:51094842	G	A	missense	*ATL1*	c.1213G>A	p.V405M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.008%	0.005%	0.020%	0.012%	0.004%	0.007%	19.5	5.33E-05	Uncertain Significance	Tolerated	0.14	Benign	
exm1102360	14:53518639	C	A	missense	*DDHD1*	c.2437+2517 G>T c.2444G>T c.2458+2517 G>T	p.R815I	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.092%	0.140%	0.000%	0.066%	0.002%	25.8	8.09E-03	Uncertain Significance	Damaging	0.04	Possibly Damaging	
exm1102387	14:53529754	T	C	missense	*DDHD1*	c.1694A>G c.1673A>G	p.Q558R p.Q565R	-	0	0	0	0	0	1	0	0	0	0	0	0	1	0	0	0	0.318%	0.170%	0.000%	0.000%	39.81	21.29	0.00	0.008%	0.007%	0.000%	0.000%	0.006%	0.007%	13.22	4.08E-05	Uncertain Significance	Damaging	0.02	Benign	
exm1102404	14:53558575	G	A	missense	*DDHD1*	c.1238C>T c.1217C>T	p.T406I p.T413I	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.001%	0.000%	0.000%	0.001%	0.000%	22.7	5.13E-05	Uncertain Significance	Damaging	0.01	Benign	
exm1102409	14:53560055	G	T	missense	*DDHD1*	c.1141C>A c.1120C>A	p.Q374K p.Q381K	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	1.85	0.024%	0.015%	0.000%	0.047%	0.015%	0.025%	22	7.55E-07	Uncertain Significance	Tolerated	0.12	Benign	
exm1102418	14:53570437	C	G	missense	*DDHD1*	c.976G>C	p.D326H	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.040%	0.026%	0.000%	0.070%	0.028%	0.044%	31	2.36E-06	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm1109904	14:68219084	C	T	missense	*ZFYVE26*	c.7348G>A	p.A2450T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.002%	0.021%	0.020%	0.012%	0.014%	0.001%	22.4	1.73E-03	Uncertain Significance	Tolerated	0.32	Benign	
exm1109913	14:68220861	G	A	missense	*ZFYVE26*	c.7055C>T	p.T2352I	-	1	3	3	0	3	1	4	0	0	1	5	1	1	0	1	0	1.592%	0.852%	0.578%	0.101%	4.28	2.29	1.55	0.372%	0.255%	0.180%	0.360%	0.247%	0.367%	23.8	6.86E-04	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm1109931	14:68222802	G	C	missense	*ZFYVE26*	c.6649C>G	p.L2217V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.015%	0.060%	0.000%	0.012%	0.000%	26.5	5.83E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm1109939	14:68228249	G	A	missense	*ZFYVE26*	c.6422C>T	p.T2141M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.008%	0.020%	0.000%	0.008%	0.002%	25.7	2.95E-04	Uncertain Significance	Damaging	0.02	Probably Damaging	
exm1109947	14:68229060	C	T	missense	*ZFYVE26*	c.6229G>A	p.G2077R	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.011%	0.198%	0.619%	0.012%	0.161%	0.010%	33	7.64E-07	Benign	Damaging	0.01	Probably Damaging	
exm1109950	14:68229095	G	A	missense	*ZFYVE26*	c.6194C>T	p.A2065V	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	3.42	0.013%	0.010%	0.000%	0.000%	0.008%	0.010%	14.53	1.59E-03	Uncertain Significance	Tolerated	0.64	Benign	
exm1109952	14:68229393	A	G	missense	*ZFYVE26*	c.6155T>C	p.V2052A	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	NaN	NaN	NaN	0.000%	0.012%	0.040%	0.000%	0.010%	0.000%	29.5	7.35E-06	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm1109955	14:68229462	A	G	missense	*ZFYVE26*	c.6086T>C	p.I2029T	-	0	2	3	1	0	1	1	1	0	2	2	3	0	1	0	0	0.000%	1.022%	0.267%	0.507%	0.00	4.33	1.13	0.236%	0.147%	0.100%	0.198%	0.133%	0.221%	25.5	8.30E-06	Uncertain Significance	Damaging	0	Probably Damaging	
exm1109960	14:68229529	T	C	missense	*ZFYVE26*	c.6019A>G	p.S2007G	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.020%	0.140%	0.000%	0.020%	0.000%	24.4	8.30E-06	Uncertain Significance	Tolerated	0.07	Probably Damaging	
exm1110016	14:68242706	C	T	missense	*ZFYVE26*	c.5092G>A	p.V1698M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.005%	0.040%	0.000%	0.008%	0.002%	27.5	1.07E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1110055	14:68249613	T	A	missense	*ZFYVE26*	c.4256A>T	p.E1419V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.031%	0.019%	0.000%	0.023%	0.015%	0.028%	25.2	7.80E-03	Uncertain Significance	Damaging	0.01	Possibly Damaging	
exm1110069	14:68249896	G	A	missense	*ZFYVE26*	c.3973C>T	p.P1325S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.007%	0.008%	0.000%	0.012%	0.008%	0.007%	16.08	3.60E-03	Uncertain Significance	Tolerated	0.1	Benign	
exm1110071	14:68249902	C	T	missense	*ZFYVE26*	c.3967G>A	p.A1323T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.001%	0.000%	0.000%	0.002%	0.002%	23.5	1.19E-04	Uncertain Significance	Tolerated	0.11	Benign	
exm1110083_ver3	14:68250174	C	T	missense	*ZFYVE26*	c.3695G>A	p.C1232Y	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.022%	0.015%	0.000%	0.058%	0.015%	0.022%	31	2.42E-06	Uncertain Significance	Damaging	0	Probably Damaging	
exm1110103	14:68252707	G	A	missense	*ZFYVE26*	c.3172C>T	p.R1058W	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.004%	0.004%	0.020%	0.000%	0.004%	0.003%	31	4.99E-03	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm1110143	14:68260350	C	T	missense	*ZFYVE26*	c.2528G>A	p.R843H	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.008%	0.000%	0.035%	0.006%	0.001%	29.4	5.53E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1110156	14:68260903	T	C	missense	*ZFYVE26*	c.2386A>G	p.S796G	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.002%	0.000%	0.012%	0.002%	0.001%	26.9	1.88E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1110190	14:68268973	G	A	missense	*ZFYVE26*	c.1462C>T	p.H488Y	-	0	2	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.341%	0.000%	0.000%	0.00	113.57	0.00	0.003%	0.002%	0.000%	0.023%	0.001%	0.002%	23.6	8.02E-03	Uncertain Significance	-	-	Possibly Damaging	
exm1110191	14:68270884	G	C	missense	*ZFYVE26*	c.1369C>G	p.L457V	-	0	0	1	0	0	0	1	1	0	0	0	0	0	0	0	0	0.000%	0.000%	0.089%	0.101%	0.00	0.00	3.56	0.025%	0.015%	0.000%	0.023%	0.016%	0.024%	26.3	4.68E-04	Uncertain Significance	Damaging	0.01	Probably Damaging	

exm1110193	14:68270902	G	A	missense	*ZFYVE26*	c.1351C>T	p.L451F	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.045%	0.028%	0.000%	0.012%	0.021%	0.036%	25.2	1.24E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm1110205	14:68272263	G	A	missense	*ZFYVE26*	c.1090C>T	p.P364S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.008%	0.006%	0.000%	0.000%	0.004%	0.007%	25.3	1.98E-06	Uncertain Significance	Damaging	0.03	Probably Damaging	
exm1110220	14:68274174	C	T	missense	*ZFYVE26*	c.827G>A	p.G276D	-	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	6.35	0.007%	0.004%	0.000%	0.012%	0.004%	0.007%	25.9	4.63E-05	Uncertain Significance	Damaging	0.02	Possibly Damaging	
exm1110256_ver4	14:68280745	C	T	stopgain	*ZFYVE26*	c.231G>A	p.W77*	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.001%	0.000%	0.012%	0.001%	0.001%	37	5.14E-05	Uncertain Significance	-	-	-	
exm1128932	14:102881115	C	T	missense	*TECPR2*	c.623C>T	p.T208I	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.049%	0.180%	0.012%	0.042%	0.000%	28.6	2.50E-04	Uncertain Significance	Damaging	0	Possibly Damaging	
exm1128979	14:102900631	C	T	missense	*TECPR2*	c.1477C>T	p.P493S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.032%	0.060%	0.012%	0.026%	0.001%	10.82	3.58E-03	Uncertain Significance	Tolerated	0.7	Benign	
exm1129060	14:102912175	C	T	missense	*TECPR2*	c.2966C>T	p.T989M	-	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	2.47	0.018%	0.023%	0.000%	0.035%	0.021%	0.018%	24.9	9.64E-07	Uncertain Significance	Damaging	0	Possibly Damaging	
exm1129073	14:102916165	C	T	missense	*TECPR2*	c.3275C>T	p.S1092L	-	0	5	16	1	4	4	7	5	3	2	3	6	1	1	2	1	2.548%	2.044%	1.246%	1.317%	4.49	3.61	2.20	0.567%	0.411%	0.120%	0.686%	0.422%	0.609%	28	2.11E-06	Uncertain Significance	Damaging	0	Probably Damaging	
exm1129076	14:102916188	G	T	missense	*TECPR2*	c.3298G>T	p.A1100S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.001%	0.000%	0.000%	0.001%	0.001%	19.92	1.09E-04	Uncertain Significance	Tolerated	0.12	Benign	
exm1129079	14:102916966	C	A	missense	*TECPR2*	c.3386C>A	p.S1129Y	-	0	0	1	0	0	0	1	0	0	0	1	0	0	0	3	0	0.000%	0.000%	0.267%	0.000%	0.00	0.00	8.09	0.033%	0.023%	0.000%	0.000%	0.027%	0.036%	30	3.15E-06	Uncertain Significance	Damaging	0	Probably Damaging	
exm1129104	14:102963323	G	T	missense	*TECPR2*	c.3797G>T	p.G1266V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.014%	0.008%	0.000%	0.035%	0.009%	0.017%	25.9	4.27E-06	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm1129115	14:102963966	G	A	missense	*TECPR2*	c.3991G>A	p.G1331R	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.004%	0.004%	0.000%	0.023%	0.002%	0.003%	33	1.00E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1129118	14:102964008	G	A	missense	*TECPR2*	c.4033G>A	p.A1345T	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	11.12	0.004%	0.138%	0.220%	0.012%	0.125%	0.005%	26.4	2.52E-06	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm1803846	14:102904413	G	A	missense	*TECPR2*	c.2449G>A	p.V817M	-	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0.000%	0.000%	0.000%	0.101%	NaN	NaN	NaN	0.000%	0.061%	0.120%	0.000%	0.061%	0.000%	23.3	4.06E-05	Uncertain Significance	Tolerated	0.06	Benign	
exm1818028_ver3	14:68241828	G	C	missense	*ZFYVE26*	c.5225C>G	p.S1742C	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.043%	0.100%	0.000%	0.055%	0.000%	28.9	5.20E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm1818043	14:68249803	A	C	missense	*ZFYVE26*	c.4066T>G	p.C1356G	-	0	0	0	0	0	0	0	0	0	0	0	2	0	0	0	0	0.000%	0.000%	0.000%	0.203%	0.00	0.00	0.00	0.003%	0.189%	0.419%	0.000%	0.222%	0.002%	22.7	7.35E-03	Benign	Tolerated	0.18	Benign	
exm2274689	14:53513535	T	C	missense	*DDHD1*	c.2570A>G c.2654A>G c.2591A>G	p.Y885C p.Y857C p.Y864C	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.016%	0.040%	0.000%	0.018%	0.000%	27.1	1.37E-05	Uncertain Significance	Damaging	0	Probably Damaging	
exm1141526_ver3	15:23060899	A	C	missense	*NIPA1*	c.233T>G c.8T>G	p.V78G p.V3G	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.094%	0.071%	0.000%	0.000%	0.051%	0.075%	23.9	4.37E-05	Uncertain Significance	Tolerated	0.07	Benign	
exm1157476	15:44855500	C	G	Splice Site	*SPG11*	c.7152-1G>C c.6813-1G>C	-	SPG11	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.035%	0.021%	0.000%	0.012%	0.016%	0.029%	25.5	2.25E-05	Likely Pathogenic	-	-	-	https://www.omim.org/entry/610844
exm1157481	15:44856827	G	A	missense	*SPG11*	c.6730C>T c.7069C>T	p.L2357F p.L2244F	-	1	0	5	0	2	1	2	0	1	0	0	1	0	1	0	2	1.274%	0.341%	0.311%	0.304%	7.36	1.97	1.80	0.173%	0.131%	0.180%	0.221%	0.144%	0.177%	25	1.12E-03	Uncertain Significance	Tolerated	0.06	Possibly Damaging	
exm1157493	15:44858173	C	T	missense	*SPG11*	c.6878G>A c.6539G>A	p.R2293Q p.R2180Q	-	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0.318%	0.000%	0.000%	0.000%	13.85	0.00	0.00	0.023%	0.017%	0.020%	0.023%	0.016%	0.026%	34	1.33E-03	Uncertain Significance	Damaging	0.01	Benign	
exm1157494	15:44858174	G	A	missense	*SPG11*	c.6877C>T c.6538C>T	p.R2293W p.R2180W	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	2.02	0.022%	0.065%	0.040%	0.012%	0.062%	0.020%	34	1.98E-03	Uncertain Significance	Damaging	0	Probably Damaging	
exm1157521	15:44862776	C	T	missense	*SPG11*	c.6085G>A c.6424G>A	p.A2142T p.A2029T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.007%	0.009%	0.040%	0.000%	0.009%	0.007%	34	6.56E-05	Uncertain Significance	Damaging	0.02	Probably Damaging	
exm1157524	15:44864905	C	T	missense	*SPG11*	c.6319G>A c.5980G>A	p.V2107I p.V1994I	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.007%	0.252%	0.759%	0.012%	0.224%	0.009%	34	1.27E-05	Benign	Damaging	0	Benign	
exm1157566	15:44877834	C	A	missense	*SPG11*	c.5121G>T	p.E1707D	-	0	1	1	0	0	0	0	0	1	1	0	1	0	0	1	0	0.318%	0.341%	0.089%	0.101%	3.93	4.21	1.10	0.081%	0.065%	0.060%	0.116%	0.064%	0.090%	23.5	1.88E-05	Uncertain Significance	Damaging	0.04	Possibly Damaging	
exm1157582	15:44884585	T	C	missense	*SPG11*	c.4687A>G	p.R1563G	-	0	0	1	0	0	0	0	0	0	0	0	1	0	0	0	0	0.000%	0.000%	0.044%	0.101%	NaN	NaN	Inf	0.000%	0.168%	0.699%	0.000%	0.153%	0.001%	24.6	6.21E-05	Benign	Damaging	0	Benign	
exm1157619	15:44900670	G	C	missense	*SPG11*	c.3425C>G	p.S1142C	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.016%	0.011%	0.000%	0.012%	0.013%	0.015%	17.08	4.73E-03	Uncertain Significance	Tolerated	0.06	Possibly Damaging	
exm1157647	15:44914006	C	A	missense	*SPG11*	c.2571G>T	p.W857C	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.000%	0.000%	0.012%	0.000%	0.000%	33	1.35E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1157656	15:44914544	A	C	missense	*SPG11*	c.2318T>G	p.V773G	-	0	0	0	0	0	0	0	0	0	0	1	1	0	0	0	0	0.000%	0.000%	0.044%	0.101%	0.00	0.00	4.04	0.011%	0.008%	0.020%	0.023%	0.008%	0.014%	28.4	7.59E-04	Uncertain Significance	Damaging	0	Probably Damaging	
exm1157716	15:44944060	C	T	stopgain	*SPG11*	c.1085G>A	p.W362*	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.001%	0.000%	0.023%	0.002%	0.003%	37	1.07E-04	Pathogenic	-	-	-	
exm1157720	15:44944355	G	C	missense	*SPG11*	c.979C>G	p.L327V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.023%	0.015%	0.020%	0.000%	0.012%	0.020%	10.17	7.98E-04	Uncertain Significance	Tolerated	0.15	Benign	
exm1157744	15:44951453	G	A	missense	*SPG11*	c.491C>T	p.S164L	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.048%	0.034%	0.000%	0.047%	0.036%	0.043%	25	3.68E-03	Uncertain Significance	Tolerated	0.17	Benign	
exm1157745	15:44951501	C	T	missense	*SPG11*	c.443G>A	p.S148N	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.013%	0.060%	0.012%	0.012%	0.002%	24.9	9.98E-05	Uncertain Significance	Tolerated	0.58	Benign	
exm1161363	15:51201108	G	T	missense	*AP4E1*	c.133G>T c.-116G>T	p.A45S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.001%	0.000%	0.000%	0.001%	0.000%	21	1.85E-03	Uncertain Significance	Tolerated	0.21	Benign	
exm1161386	15:51221276	C	A	missense	*AP4E1*	c.613C>A c.388C>A	p.H130N p.H205N	-	0	0	1	0	1	0	0	1	0	0	1	0	0	0	0	0	0.318%	0.000%	0.089%	0.101%	5.79	0.00	1.62	0.055%	0.033%	0.020%	0.058%	0.038%	0.065%	32	3.66E-06	Uncertain Significance	Tolerated	0.1	Possibly Damaging	
exm1161417	15:51240316	A	C	missense	*AP4E1*	c.1051A>C c.1276A>C	p.I351L p.I426L	-	0	0	3	0	1	0	4	3	0	2	4	1	0	0	0	1	0.318%	0.341%	0.489%	0.507%	1.97	2.10	3.02	0.162%	0.105%	0.100%	0.116%	0.107%	0.167%	24.7	2.38E-05	Uncertain Significance	Tolerated	0.27	Possibly Damaging	
exm1161443	15:51260460	G	A	missense	*AP4E1*	c.1627G>A c.1852G>A	p.V543I p.V618I	-	0	0	1	0	0	0	1	0	0	0	0	0	0	0	0	1	0.000%	0.000%	0.089%	0.101%	0.00	0.00	1.51	0.059%	0.046%	0.020%	0.047%	0.042%	0.059%	26.6	2.94E-05	Uncertain Significance	Damaging	0.02	Benign	
exm1169606	15:65262475	C	T	missense	*SPG21*	c.457G>A c.538G>A	p.A180T p.A153T	-	0	1	2	0	0	3	1	0	1	0	2	2	1	1	0	0	0.637%	0.852%	0.222%	0.203%	4.94	6.60	1.72	0.129%	0.097%	0.000%	0.233%	0.091%	0.122%	34	9.38E-07	Uncertain Significance	Damaging	0.05	Benign	
exm1831842	15:44864940	A	G	missense	*SPG11*	c.5945T>C c.6284T>C	p.L1982S p.L2095S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.070%	0.180%	0.000%	0.078%	0.000%	27.5	2.54E-03	Uncertain Significance	Damaging	0.05	Possibly Damaging	
exm1833697	15:51223090	A	G	missense	*AP4E1*	c.566A>G c.791A>G	p.N264S p.N189S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.004%	0.059%	0.160%	0.000%	0.069%	0.003%	14.14	5.14E-03	Uncertain Significance	Tolerated	0.17	Benign	
exm1833716	15:51250834	C	A	missense	*AP4E1*	c.1694C>A c.1469C>A	p.A490E p.A565E	-	0	0	1	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.089%	0.000%	NaN	NaN	Inf	0.000%	0.020%	0.000%	0.000%	0.017%	0.000%	19.26	1.05E-04	Uncertain Significance	Tolerated	0.2	Benign	
exm1833737	15:51293344	C	A	missense	*AP4E1*	c.2992C>A c.3217C>A	p.Q1073K p.Q998K	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.066%	0.160%	0.000%	0.070%	0.001%	11.55	4.90E-03	Uncertain Significance	Tolerated	1	Benign	
exm1837123	15:65273363	T	C	missense	*SPG21*	c.64A>G	p.I22V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.031%	0.080%	0.000%	0.029%	0.000%	13.13	4.56E-04	Uncertain Significance	Tolerated	0.36	Benign	
exm1258691	16:74761247	A	C	missense	*FA2H*	c.401T>G	p.V134G	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.835%	0.793%	0.000%	0.000%	0.624%	0.715%	23.5	3.38E-04	Uncertain Significance	Damaging	0	Benign	
exm1270646	16:89597107	C	T	missense	*SPG7*	c.878C>T	p.A293V	-	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	NaN	NaN	Inf	0.000%	0.001%	0.020%	0.000%	0.001%	0.000%	31	5.20E-07	Uncertain Significance	Tolerated	0.06	Probably Damaging	
exm1270647_ver4	16:89597110	G	A	missense	*SPG7*	c.881G>A	p.R294H	-	0	0	1	0	0	0	0	1	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.101%	0.00	0.00	8.90	0.005%	0.339%	0.998%	0.012%	0.295%	0.007%	27.9	1.04E-03	Benign	Tolerated	0.08	Possibly Damaging	
exm1270656	16:89598369	G	A	missense	*SPG7*	c.1045G>A	p.G349S	SPG7	0	1	4	0	0	1	6	2	0	0	1	4	0	0	0	0	0.000%	0.341%	0.489%	0.608%	0.00	2.60	3.74	0.131%	0.086%	0.080%	0.174%	0.091%	0.144%	31	5.25E-07	Likely Pathogenic	Damaging	0	Probably Damaging	https://www.omim.org/entry/602783
exm1270668	16:89598987	G	A	missense	*SPG7*	c.1267G>A	p.G423S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.008%	0.020%	0.012%	0.011%	0.001%	22.9	5.51E-04	Uncertain Significance	Tolerated	0.1	Benign	
exm1270714	16:89613145	C	T	missense	*SPG7*	c.1529C>T	p.A510V	SPG7	1	1	8	0	5	5	2	4	2	1	9	7	0	2	1	2	2.548%	1.533%	0.890%	1.317%	6.87	4.13	2.40	0.371%	0.252%	0.220%	0.453%	0.266%	0.416%	32	8.91E-07	Likely Pathogenic	Damaging	0	Probably Damaging	https://www.omim.org/entry/602783
exm1270715	16:89613169	G	T	Splice Site	*SPG7*	c.1552+1G>T	-	SPG7	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.318%	0.000%	0.000%	0.000%	63.69	0.00	0.00	0.005%	0.003%	0.000%	0.023%	0.004%	0.006%	23.9	8.91E-07	Likely Pathogenic	-	-	-	https://www.ncbi.nlm.nih.gov/pubmed/20108356
exm1270744	16:89620204	G	T	missense	*SPG7*	c.1939G>T	p.A647S	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.008%	0.007%	0.000%	0.047%	0.007%	0.009%	26	5.93E-06	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm1873000	16:89592774	T	C	missense	*SPG7*	c.656T>C	p.I219T	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.029%	0.120%	0.000%	0.032%	0.000%	23.6	1.05E-05	Uncertain Significance	Tolerated	0.11	Benign	
exm1416146	19:7605870	G	C	missense	*PNPLA6*	c.857G>C c.884G>C c.740G>C	p.R247P p.R295P p.R286P	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.000%	0.000%	0.000%	0.003%	0.006%	27.9	1.30E-04	Uncertain Significance	Damaging	0.02	Probably Damaging	
exm1416186	19:7607741	C	T	missense	*PNPLA6*	c.1340C>T c.1457C>T c.1484C>T	p.P486L p.P495L p.P447L	-	1	1	6	3	3	6	5	3	0	1	5	5	0	1	2	1	1.274%	1.533%	0.801%	1.216%	1.52	1.83	0.95	0.839%	0.548%	0.359%	0.687%	0.522%	0.841%	22.8	1.19E-04	Uncertain Significance	Tolerated	0.39	Benign	
exm1416240	19:7620171	G	C	missense	*PNPLA6*	c.2659G>C c.2740G>C c.2854G>C c.2884G>C	p.D962H p.D952H p.D914H p.D887H	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.026%	0.000%	0.000%	0.019%	0.000%	27.8	6.76E-06	Uncertain Significance	Tolerated	0.17	Probably Damaging	
exm1416245	19:7620495	G	C	missense	*PNPLA6*	c.2744G>C c.2969G>C c.2825G>C c.2939G>C	p.G915A p.G980A p.G990A p.G942A	-	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.044%	0.000%	0.00	0.00	1.39	0.032%	0.019%	0.000%	0.012%	0.019%	0.033%	25.5	8.51E-06	Uncertain Significance	Damaging	0.01	Probably Damaging	
exm1416269	19:7623766	C	T	missense	*PNPLA6*	c.3233C>T c.3314C>T c.3428C>T c.3458C>T	p.T1105M p.T1143M p.T1153M p.T1078M	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.003%	0.017%	0.020%	0.012%	0.013%	0.002%	23.1	2.38E-03	Uncertain Significance	Tolerated	0.25	Possibly Damaging	
exm1480937	19:45988997	G	A	missense	*RTN2*	c.1607C>T c.1388C>T c.587C>T	p.A536V p.A196V p.A463V	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	NaN	NaN	NaN	0.000%	0.007%	0.000%	0.000%	0.007%	0.000%	23.7	1.55E-03	Uncertain Significance	Tolerated	0.12	Benign	
exm1481012	19:45998398	C	T	Splice Site	*RTN2*	c.35-1G>A	-	-	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.000%	0.000%	0.000%	0.000%	0.00	0.00	0.00	0.001%	0.001%	0.000%	0.012%	0.001%	0.001%	25.8	9.68E-04	Uncertain Significance	-	-	-	