Supplementary Table 1. Variants identified in the 11 genes responsible for non-syndromic eoHM based on our cohort.

chr#	Gene	Position	Nucleotide Change	Effect	sample count	Diagnosis		gnomAD Allele		SIFT	PROVEAN	Polyphen-2	CADD	REVEL	HGMD
						eoHM	Others	Count	Frequency						
chr22	*SCO2*	50962077	c.764G>A	p.R255Q	1	0	1	11	3.89E-05	T(0.565)	N(-0.62)	B(0.053)	11.16	0.235	
chr22	*SCO2*	50962089	c.752C>T	p.S251L	29	4	25	48	0.000169785	D(0.028)	N(-2.16)	B(0.05)	15.17	0.148	
chr22	*SCO2*	50962117	c.724G>A	p.G242S	1	0	1	1	3.18E-05	T(0.051)	D(-4.69)	P(0.758)	15.93	0.369	
chr22	*SCO2*	50962128	c.713C>T	p.T238M	2	0	2	37	0.000130835	T(0.088)	N(1.17)	P(0.512)	22.6	0.401	
chr22	*SCO2*	50962165	c.676A>G	p.I226V	1	0	1	1	3.97722E-06	T(0.067)	N(-0.78)	B(0.026)	16.4	0.326	
chr22	*SCO2*	50962177	c.664G>A	p.V222M	2	0	2	9	3.18226E-05	D(0.001)	D(-2.88)	D(0.994)	26.3	0.774	
chr22	*SCO2*	50962222	c.619G>A	p.V207M	1	0	1	14	4.9505E-05	D(0)	D(-2.94)	D(0.999)	24	0.802	
chr22	*SCO2*	50962264	c.577G>A	p.G193S	1	0	1	8	3.18281E-05	D(0)	D(-5.89)	D(1)	24.1	0.831	
chr22	*SCO2*	50962300	c.541G>A	p.V181I	5	1	4	22	7.78243E-05	T(0.165)	N(-0.64)	B(0.262)	15.42	0.474	
chr22	*SCO2*	50962306	c.535C>T	p.R179C	1	0	1	13	4.59936E-05	D(0.004)	D(-4.68)	D(0.969)	24.6	0.639	
chr22	*SCO2*	50962321	c.520G>A	p.V174I	2	0	2	2	7.96248E-06	T(0.245)	N(-0.86)	B(0.014)	13.79	0.388	
chr22	*SCO2*	50962395	c.446G>A	p.R149Q	1	0	1	14	4.95642E-05	T(0.263)	N(1.04)	B(0.005)	6.637	0.16	
chr22	*SCO2*	50962434	c.407T>C	p.I136T	2	2	0	/	/	D(0.001)	D(-4.48)	D(0.99)	26.2	0.937	
chr22	*SCO2*	50962455	c.386G>T	p.G129V	3	1	2	1	3.98206E-06	D(0)	D(-8.44)	D(1)	26.8	0.82	
chr22	*SCO2*	50962458	c.383T>A	p.F128Y	1	0	1	/	/	D(0)	D(-2.89)	D(0.998)	27	0.929	
chr22	*SCO2*	50962482	c.359G>A	p.R120Q	1	0	1	6	2.12681E-05	T(0.113)	N(-0.37)	B(0.021)	19.59	0.262	
chr22	*SCO2*	50962483	c.358C>T	p.R120W	3	1	2	1	3.98765E-06	D(0.04)	N(-1.17)	P(0.881)	26.7	0.83	
chr22	*SCO2*	50962492	c.349G>A	p.A117T	1	0	1	/	/	T(0.239)	N(-0.2)	B(0.333)	18.92	0.225	
chr22	*SCO2*	50962500	c.341G>A	p.R114H	1	0	1	212	0.000752269	D(0.019)	D(-3.47)	D(0.958)	25.7	0.62	Khanh-Nhat TV, et al.
chr22	*SCO2*	50962507	c.334C>T	p.R112W	7	1	6	14	4.96789E-05	D(0.003)	D(-3.52)	P(0.567)	23	0.74	
chr22	*SCO2*	50962514	c.327C>G	p.H109Q	1	0	1	6	2.39674E-05	D(0.049)	D(-4.57)	P(0.51)	14.33	0.365	

chr22	*SCO2*	50962549	c.292G>T	p.A98S	1	0	1	1	4.01532E-06	T(0.18)	N(-1.88)	B(0.034)	10.03	0.124	Tomotaka W, et al.
chr22	*SCO2*	50962551	c.290C>T	p.A97V	7	0	7	7	2.81253E-05	T(0.35)	N(-1.75)	P(0.679)	17.42	0.306	
chr22	*SCO2*	50962612	c.210_229del	p.L71Pfs*4	2	0	2	/	/	NA	NA	NA	NA	NA	
chr22	*SCO2*	50962627	c.214G>A	p.G72S	5	0	5	7	2.55199E-05	D(0.0490	N(0.3)	B(0.045)	9.33	0.215	
chr22	*SCO2*	50962652	c.189C>G	p.I63M	2	0	2	2	8.25962E-06	D(0.032)	N(-1.29)	B(0.146)	13.35	0.223	
chr22	*SCO2*	50962719	c.122A>C	p.Q41P	1	0	1	1	3.1953E-05	T(0.079)	N(0.1)	B(0.001)	8.088	0.26	
chr22	*SCO2*	50962786	c.55C>T	p.P19S	2	0	2	1	4.24203E-06	D(0.035)	N(0.45)	B(0.001)	2.265	0.132	
chr22	*SCO2*	50962802	c.39delG	p.R13Sfs*21	1	0	1	/	/	NA	NA	NA	NA	NA	
chr01	*ZNF644*	91382402	c.3937A>G	p.I1313V	1	0	1	34	0.000120207	D(0.002)	N(-0.14)	P(0.748)	23.1	0.187	
chr01	*ZNF644*	91383615	c.3785G>A	p.R1262Q	1	0	1	1	3.18431E-05	D(0.006)	N(-1.57)	D(0.919)	26	0.311	
chr01	*ZNF644*	91403051	c.3679A>G	p.M1227V	1	0	1	8	2.83531E-05	T(0.122)	N(-0.57)	B(0)	15.97	0.168	
chr01	*ZNF644*	91403111	c.3619G>A	p.V1207I	1	0	1	12	4.25786E-05	T(0.077)	N(-0.24)	B(0.191)	19.01	0.152	
chr01	*ZNF644*	91403179	c.3551G>T	p.R1184M	8	1	7	13	5.18763E-05	T(0.072)	N(-0.31)	B(0.006)	21.8	0.229	
chr01	*ZNF644*	91403191	c.3539T>C	p.M1180T	1	0	1	/	/	D(0.008)	N(-0.29)	B(0.014)	22.2	0.208	
chr01	*ZNF644*	91403435	c.3295C>T	p.R1099C	1	1	0	9	3.18961E-05	D(0.0)	N(-2)	D(0.993)	30	0.252	Zheng YH, et al. Cai XB, et al.
chr01	*ZNF644*	91403464	c.3266A>G	p.Y1089C	33	1	32	65	0.000230216	D(0.0)	N(-2.24)	D(0.982)	24.4	0.235	
chr01	*ZNF644*	91403469	c.3261A>C	p.E1087D	1	0	1	/	/	D(0.007)	N(-0.89)	B(0.001)	19.17	0.338	
chr01	*ZNF644*	91403597	c.3133G>T	p.G1045C	2	0	2	/	/	D(0.0)	N(-1.66)	D(0.996)	31	0.192	
chr01	*ZNF644*	91403626	c.3104C>A	p.T1035N	1	1	0	/	/	T(0.137)	N(-0.53)	B(0.026)	21.7	0.064	
chr01	*ZNF644*	91404080	c.2831C>T	p.A944V	1	1	0	3	1.19822E-05	D(0.001)	N(-0.79)	B(0.001)	21.5	0.183	
chr01	*ZNF644*	91404107	c.2804A>G	p.D935G	1	0	1	1	3.99642E-06	T(0.148)	N(-1.1)	B(0.018)	22.0	0.201	
chr01	*ZNF644*	91404126	c.2785T>A	p.F929I	1	1	0	/	/	D(0.001)	N(-0.49)	B(0.205)	23.8	0.237	
chr01	*ZNF644*	91404178	c.2733C>A	p.D911E	1	0	1	/	/	T(0.46)	N(0.33)	B(0.001)	12.53	0.212	
chr01	*ZNF644*	91404273	c.2638A>G	p.S880G	1	0	1	/	/	T(0.218)	N(-0.36)	B(0)	15.34	0.072	

chr01	*ZNF644*	91404360	c.2551G>C	p.D851H	1	1	0	/	/	D(0.0)	N(-1.45)	P(0.573)	25.1	0.662	
chr01	*ZNF644*	91404408	c.2503A>G	p.K835Q	1	1	0	/	/	D(0.0)	N(-0.95)	P(0.823)	26.1	0.23	
chr01	*ZNF644*	91404438	c.2473G>T	p.D825Y	1	0	1	/	/	D(0.001)	N(-1.18)	B(0.34)	22.9	0.085	
chr01	*ZNF644*	91404576	c.2335A>G	p.S779G	1	1	0	26	9.20354E-05	T(0.097)	N(-0.62)	B(0)	15.21	0.039	
chr01	*ZNF644*	91404576	c.2332_2334del	p.Ser779del	1	0	1	3	1.06195E-05	NA	NA	NA	NA	NA	
chr01	*ZNF644*	91404614	c.2297C>A	p.A766D	1	0	1	/	/	D(0.001)	N(-0.53)	B(0.001)	15.64	0.141	
chr01	*ZNF644*	91404614	c.2294_2296del	p.Glu765del	1	0	1	19	6.72986E-05	NA	NA	NA	NA	NA	
chr01	*ZNF644*	91404630	c.2281T>C	p.F761L	1	1	0	/	/	T(0.55)	N(0.87)	B(0)	17.92	0.236	
chr01	*ZNF644*	91404725	c.2186A>G	p.K729R	1	1	0	/	/	D(0.005)	N(-0.36)	B(0.009)	22.2	0.225	
chr01	*ZNF644*	91404749	c.2162A>G	p.Y721C	4	1	3	11	3.89568E-05	D(0.0)	N(-1.03)	B(0.01)	22.5	0.142	
chr01	*ZNF644*	91404780	c.2131A>G	p.I711V	1	0	1	64	0.000226643	T(0.627)	N(0.2)	B(0)	0.001	0.03	
chr01	*ZNF644*	91404783	c.2128A>G	p.T710A	1	0	1	/	/	T(0.091)	N(0.23)	B(0.002)	2.965	0.031	
chr01	*ZNF644*	91404794	c.2117A>G	p.H706R	1	0	1	/	/	T(0.112)	N(-0.15)	B(0)	5.645	0.06	
chr01	*ZNF644*	91404863	c.2048G>C	p.R683T	1	1	0	38	0.000134702	D(0.034)	N(-0.58)	P(0.558)	24.8	0.681	
chr01	*ZNF644*	91404897	c.2014A>G	p.S672G	5	1	4	1	3.98629E-06	D(0.001)	N(-0.71)	B(0.282)	16.42	0.157	Shi Y, et al.
chr01	*ZNF644*	91404947	c.1964C>G	p.S655C	1	0	1	3	1.19619E-05	D(0.0)	N(-1.59)	D(0.993)	24.7	0.364	
chr01	*ZNF644*	91404953	c.1958A>G	p.K653R	1	0	1	4	1.59537E-05	D(0.001)	N(-0.9)	D(0.98)	24.2	0.191	
chr01	*ZNF644*	91404960	c.1951T>C	p.F651L	1	0	1	/	/	D(0.007)	N(-1.3)	D(0.952)	24.5	0.259	
chr01	*ZNF644*	91404978	c.1933A>G	p.K645E	1	0	1	1	3.18552E-05	D(0.023)	N(-0.67)	B(0.192)	22.6	0.237	
chr01	*ZNF644*	91405108	c.1803A>T	p.L601F	1	0	1	20	7.96134E-05	T(0.13)	D(-2.5)	B(0.004)	16.26	0.184	
chr01	*ZNF644*	91405152	c.1759A>G	p.I587V	1	0	1	7	2.47774E-05	T(0.646)	N(0.2)	B(0)	0.009	0.025	Shi Y, et al.
chr01	*ZNF644*	91405217	c.1694A>G	p.K565R	1	0	1	31	0.00011004	T(0.102)	N(-0.52)	D(0.98)	23.3	0.185	
chr01	*ZNF644*	91405229	c.1682T>C	p.I561T	1	0	1	/	/	T(0.079)	N(-0.74)	B(0)	2.008	0.077	
chr01	*ZNF644*	91405274	c.1637T>C	p.I546T	1	0	1	/	/	T(0.228)	N(0.16)	B(0.005)	17.88	0.133	

chr01	*ZNF644*	91405427	c.1484G>T	p.R495L	1	0	1	/	/	D(0.005)	D(-3.7)	D(0.932)	26.9	0.239	
chr01	*ZNF644*	91405508	c.1403T>C	p.I468T	1	0	1	/	/	T(0.068)	N(-0.08)	D(0.998)	24	0.439	
chr01	*ZNF644*	91405512	c.1399A>G	p.M467V	1	0	1	5	1.9911E-05	T(0.006)	N(-1.8)	D(0.977)	24.5	0.437	Cai XB, et al.
chr01	*ZNF644*	91405754	c.1157C>G	p.T386S	1	0	1	/	/	T(0.259)	N(-0.37)	B(0.001)	17.01	0.296	
chr01	*ZNF644*	91405772	c.1139G>A	p.S380N	1	0	1	/	/	T(0.232)	N(0.01)	B(0.01)	19.25	0.249	
chr01	*ZNF644*	91405782	c.1129G>A	p.V377I	1	0	1	/	/	T(0.189)	N(-0.26)	B(0.001)	21.5	0.298	
chr01	*ZNF644*	91405805	c.1106A>T	p.K369M	1	1	0	/	/	D(0.005)	N(-1.24)	P(0.453)	24.2	0.19	
chr01	*ZNF644*	91405811	c.1100C>T	p.P367L	36	2	34	23	8.13981E-05	D(0.001)	D(-2.62)	B(0.033)	23	0.159	
chr01	*ZNF644*	91405817	c.1094A>G	p.Y365C	1	0	1	1	3.98089E-06	T(0.139)	N(-1.36)	B(0.001)	19.09	0.28	
chr01	*ZNF644*	91405869	c.1042G>C	p.E348Q	1	0	1	/	/	D(0.009)	N(-0.2)	D(0.966)	23.2	0.195	
chr01	*ZNF644*	91405943	c.965_967del	p.Glu322del	4	0	4	2	7.97137E-06	NA	NA	NA	NA	NA	
chr01	*ZNF644*	91405948	c.963A>T	p.Q321H	1	0	1	/	/	D(0.022)	N(0.03)	B(0.002)	14.87	0.073	
chr01	*ZNF644*	91405979	c.932C>G	p.S311C	1	0	1	/	/	D(0.028)	N(-1.41)	P(0.533)	21.8	0.166	
chr01	*ZNF644*	91405998	c.913G>A	p.E305K	83	2	81	202	0.000716058	D(0.002)	N(-1.15)	D(0.097)	26.2	0.23	Liu F, et al.
chr01	*ZNF644*	91406007	c.904C>T	p.R302C	1	0	1	5	1.99461E-05	D(0.002)	N(-1.39)	D(0.988)	28.5	0.356	
chr01	*ZNF644*	91406082	c.829G>A	p.D277N	4	0	4	/	/	D(0.004)	N(-1.21)	B(0.205)	22.9	0.247	
chr01	*ZNF644*	91406351	c.560C>T	p.P187L	6	0	6	10	3.54313E-05	T(0.078)	N(-1.03)	B(0.001)	20.8	0.216	
chr01	*ZNF644*	91406391	c.520G>C	p.V174L	1	0	1	/	/	T(0.158)	N(-0.45)	B(0.002)	21.3	0.229	
chr01	*ZNF644*	91406417	c.494C>G	p.T165R	4	0	4	9	3.18972E-05	D(0.001)	N(-1.56)	B(0.091)	23.3	0.295	
chr01	*ZNF644*	91406495	c.416C>G	p.T139S	1	0	1	/	/	D(0.007)	N(-0.6)	P(0.901)	22.9	0.28	
chr01	*ZNF644*	91406607	c.304A>G	p.T102A	1	0	1	7	2.47884E-05	T(0.551)	N(-0.2)	B(0)	13	0.203	
chr01	*ZNF644*	91406741	c.170A>G	p.H57R	1	0	1	2	8.01462E-06	D(0.027)	N(-0.43)	B(0)	9.452	0.129	
chr01	*ZNF644*	91406799	c.112G>A	p.A38T	1	0	1	1	4.14983E-06	D(0.0)	N(-1.07)	D(0.989)	25.3	0.272	
chr01	*ZNF644*	91447882	c.29A>T	p.N10I	1	0	1	1	4.01745E-06	T(1)	N(0.91)	B(0.274)	22.5	0.047	

chr01	*ZNF644*	91447889	c.22G>T	p.D8Y	1	0	1	/	/	D(0.0)	N(-0.41)	B(0.001)	25.5	0.163	
chr04	*CCDC111*	185578298	c.4A>G	p.N2D	1	0	1	/	/	T(0.422)	N(-0.78)	B(0.016)	19.06	0.055	
chr04	*CCDC111*	185578346	c.52C>G	p.H18D	1	0	1	/	/	T(0.235)	N(-0.94)	B(0.032)	12.99	0.068	
chr04	*CCDC111*	185578374	c.80T>C	p.V27A	1	0	1	/	/	T(0.641)	N(-0.72)	B(0.152)	16.45	0.038	
chr04	*CCDC111*	185578410	c.116C>G	p.P39R	1	0	1	/	/	T(0.064)	N(-0.86)	B(0.277)	22.9	0.109	
chr04	*CCDC111*	185580539	c.226C>T	p.R76C	2	0	2	15	5.96749E-05	D(0.0)	D(-6.64)	D(1.0)	27.2	0.672	Zhao FX, et al.
chr04	*CCDC111*	185580578	c.265T>G	p.Y89D	42	1	41	122	0.000431391	D(0.009)	D(-4.19)	P(0.771)	23.6	0.265	
chr04	*CCDC111*	185580582	c.269A>G	p.Y90C	1	0	1	4	1.41451E-05	D(0.001)	D(-5.65)	D(1.0)	26.3	0.639	
chr04	*CCDC111*	185583016	c.367C>T	p.P123S	5	0	5	7	2.78567E-05	D(0.006)	D(-3.23)	P(0.756)	24.8	0.211	
chr04	*CCDC111*	185583038	c.390dupG	p.V131Gfs*6	137	16	121	276	0.000976887	NA	NA	NA	NA	NA	
chr04	*CCDC111*	185583041	c.392T>G	p.V131G	3	1	2	/	/	D(0.001)	D(-5.48)	D(1.0)	29.5	0.776	
chr04	*CCDC111*	185583044	c.395C>T	p.A132V	1	0	1	/	/	T(0.146)	N(-1.58)	B(0.208)	23.8	0.141	
chr04	*CCDC111*	185587098	c.436T>C	p.Y146H	5	1	4	6	2.43681E-05	D(0.036)	D(-4.31)	B(0.067)	17.06	0.157	
chr04	*CCDC111*	185587099	c.437A>G	p.Y146C	1	0	1	/	/	D(0.002)	D(-7.19)	D(0.916)	23	0.369	
chr04	*CCDC111*	185587140	c.478T>C	p.S160P	1	0	1	/	/	D(0.006)	D(-4.25)	P(0.898)	26.7	0.604	
chr04	*CCDC111*	185587164	c.502C>T	p.Q168*	1	0	1	16	6.39044E-05	NA	NA	NA	39	NA	
chr04	*CCDC111*	185587188	c.526G>A	p.D176N	1	0	1	/	/	T(0.755)	N(0.61)	B(0.002)	16.54	0.055	
chr04	*CCDC111*	185587215	c.553G>A	p.V185I	12	2	10	42	0.000155987	T(0.112)	N(-0.83)	B(0.026)	22.3	0.2	
chr04	*CCDC111*	185593346	c.576T>G	p.I192M	5	1	4			T(0.195)	N(-1.58)	D(0.947)	21	0.239	
chr04	*CCDC111*	185593584	c.814A>G	p.M272V	1	0	1	/	/	T(0.3)	N(-0.37)	B(0.0)	0.001	0.012	
chr04	*CCDC111*	185599423	c.882A>C	p.K294N	1	1	0	2	8.52798E-06	D(0.025)	N(-2.38)	D(0.995)	23.9	0.186	
chr04	*CCDC111*	185599443	c.902G>A	p.R301H	1	0	1	48	0.000176651	T(0.551)	N(-1.00)	T(0.001)	5.586	0.021	
chr04	*CCDC111*	185599473	c.932A>G	p.K311R	2	0	2	3	1.22054E-05	T(0.788)	N(0.02)	B(0.001)	0.001	0.077	
chr04	*CCDC111*	185599478	c.937T>G	p.F313V	1	0	1	2	8.13464E-06	T(0.639)	N(0.64)	B(0.003)	12.98	0.042	

chr04	*CCDC111*	185599544	c.1003G>A	p.V335I	1	0	1	1	4.16438E-06	T(0.126)	N(-0.36)	B(0.032)	16.62	0.033	
chr04	*CCDC111*	185603419	c.1025G>A	p.R342Q	1	0	1	1	4.01287E-06	D(0.033)	N(-2.08)	B(0.028)	23.1	0.155	
chr04	*CCDC111*	185603427	c.1033A>G	p.T345A	1	0	1	/	/	D(0.040)	N(-1.48)	B(0.137)	22.3	0.064	
chr04	*CCDC111*	185606572	c.1106T>C	p.I369T	12	0	12	30	0.000106151	T(0.31)	N(0.17)	B(0.031)	15.95	0.09	
chr04	*CCDC111*	185606598	c.1132C>A	p.P378T	1	0	1	1	3.97962E-06	D(0.009)	D(-6.03)	P(0.863)	25	0.246	
chr04	*CCDC111*	185606611	c.1145A>G	p.H382R	2	0	2	2	7.95957E-06	T(0.553)	N(0.32)	B(0.0)	5.323	0.045	
chr04	*CCDC111*	185606650	c.1184G>C	p.G395A	4	0	4	17	6.01485E-05	T(0.146)	D(-4.14)	D(0.998)	31	0.466	
chr04	*CCDC111*	185606738	c.1195C>T	p.R399C	1	0	1	4	1.41751E-05	D(0.046)	D(-2.73)	B(0.018)	23.9	0.12	
chr04	*CCDC111*	185606739	c.1196G>A	p.R399H	2	0	2	7	2.78951E-05	D(0.029)	N(-1.54)	B(0.018)	19.57	0.133	
chr04	*CCDC111*	185606778	c.1235A>G	p.D412G	1	0	1	/	/	D(0.0)	D(-4.51)	D(0.991)	29.2	0.54	
chr04	*CCDC111*	185606792	c.1249C>T	p.R417W	2	0	2	19	6.73315E-05	D(0.0)	D(-7.73)	D(1.0)	33	0.644	
chr04	*CCDC111*	185606793	c.1250G>A	p.R417Q	1	0	1	10	3.98708E-05	D(0.0)	D(-3.87)	D(0.999)	32	0.646	
chr04	*CCDC111*	185612822	c.1381T>C	p.F461L	1	1	0	2	7.96197E-06	T(0.491)	N(-1.25)	B(0.001)	20.9	0.063	
chr04	*CCDC111*	185615675	c.1431_1433delAGA	p.Glu478del	27	5	22	62	0.000220164	NA	NA	NA	NA	NA	
chr04	*CCDC111*	185615718	c.1468_1471del	p.N490Kfs*33	1	0	1	/	/	NA	NA	NA	NA	NA	
chr04	*CCDC111*	185615843	c.1593_1594insGC	p.E533Lfs*22	1	0	1	/	/	NA	NA	NA	NA	NA	
chr12	*SLC39A5*	56625089	c.31G>A	p.A11T	1	1	0	2	8.11004E-06	T(0.175)	N(-0.34)	B(0.001)	12.6	0.06	
chr12	*SLC39A5*	56625098	c.40T>C	p.C14R	1	0	1	1	4.03672E-06	D(0.004)	N(-1.83)	B(0.102)	17.07	0.17	
chr12	*SLC39A5*	56625099	c.41G>C	p.C14S	1	1	0	5	2.01475E-05	D(0.009)	N(-1.07)	B(0.002)	13.02	0.057	
chr12	*SLC39A5*	56625199	c.141C>G	p.Y47*	4	0	4	6	2.12441E-05	NA	NA	NA	28.4	NA	Guo H, et al.
chr12	*SLC39A5*	56625237	c.179G>A	p.R60Q	1	0	1	58	0.000205894	D(0)	N(-2.23)	P(0.637)	25	0.069	
chr12	*SLC39A5*	56625260	c.202G>C	p.G68R	1	0	1	/	/	D(0)	D(-6)	D(0.999)	27.3	0.393	
chr12	*SLC39A5*	56625270	c.212A>G	p.Q71R	2	0	2	1	4.03584E-06	T(0.325)	N(-0.47)	P(0.489)	24.3	0.082	
chr12	*SLC39A5*	56625308	c.250C>T	p.R84W	2	0	2	28	0.000124739	D(0.001)	N(-1.97)	P(0.635)	23.8	0.107	Cai XB, et al.

chr12	*SLC39A5*	56625309	c.251G>A	p.R84Q	2	0	2	19	7.41475E-05	T(0.06)	N(-1.24)	B(0.008)	5.113	0.016	
chr12	*SLC39A5*	56626510	c.325G>T	p.G109W	17	1	16	30	0.000107374	D(0)	D(-3.23)	P(0.868)	25.3	0.167	
chr12	*SLC39A5*	56626574	c.389G>A	p.R130H	16	0	16	45	0.000160594	T(0.586)	N(0.82)	B(0.0)	5.244	0.017	
chr12	*SLC39A5*	56626589	c.404C>T	p.S135L	10	0	10	23	8.18867E-05	D(0.007)	N(-1.75)	P(0.576)	26.7	0.051	
chr12	*SLC39A5*	56626624	c.439G>C	p.D147H	1	0	1	/	/	T(0.089)	N(-1.85)	D(0.997)	25	0.17	
chr12	*SLC39A5*	56626628	c.443A>C	p.H148P	1	0	1	/	/	D(0)	D(-5.5)	D(0.991)	25.7	0.147	
chr12	*SLC39A5*	56626657	c.471+1G>T	splicing	2	0	2	/	/	NA	NA	NA	33	NA	
chr12	*SLC39A5*	56628660	c.524C>G	p.P175R	1	0	1	/	/	T(0.248)	N(-2.05)	B(0.174)	20.9	0.212	
chr12	*SLC39A5*	56628672	c.536G>C	p.R179P	1	0	1	/	/	T(0.244)	N(-1.01)	B(0.071)	10.68	0.159	
chr12	*SLC39A5*	56628690	c.554G>A	p.C185Y	1	0	2	5	1.76921E-05	D(0)	D(-9.64)	D(0.997)	27.4	0.841	
chr12	*SLC39A5*	56628712	c.576C>G	p.I192M	1	0	1	/	/	D(0)	N(-1.88)	P(0.682)	14.93	0.489	
chr12	*SLC39A5*	56628713	c.577G>A	p.D193N	7	0	7	24	8.49642E-05	D(0.002)	D(-3.91)	P(0.533)	25.8	0.382	
chr12	*SLC39A5*	56628722	c.586G>A	p.V196I	1	0	1	18	7.16994E-05	T(0.116)	N(-0.66)	P(0.887)	23.5	0.237	
chr12	*SLC39A5*	56628738	c.602C>T	p.P201L	1	0	1	18	6.37945E-05	T(0.763)	N(-0.83)	B(0.002)	12.47	0.052	
chr12	*SLC39A5*	56628954	c.648T>G	p.S216R	2	0	2	1	5.07882E-06	D(0.003)	D(-2.65)	B(0.158)	17.77	0.322	
chr12	*SLC39A5*	56629010	c.704G>A	p.R235Q	2	0	2	10	3.90183E-05	D(0.023)	N(0.21)	D(0.914)	23.7	0.261	
chr12	*SLC39A5*	56629025	c.719G>A	p.R240H	1	0	1	3	1.33407E-05	T(0.192)	N(-0.35)	B(0.003)	11.51	0.055	
chr12	*SLC39A5*	56629033	c.727C>T	p.R243W	1	0	1	15	5.94036E-05	D(0.001)	D(-3.65)	P(0.88)	25.5	0.302	
chr12	*SLC39A5*	56629055	c.749G>A	p.G250E	1	1	0	/	/	T(0.082)	N(-0.38)	D(0.958)	24.1	0.236	
chr12	*SLC39A5*	56629064	c.758C>A	p.A253E	1	0	1	3	1.54316E-05	D(0)	D(-4.68)	D(1.0)	32	0.821	
chr12	*SLC39A5*	56629362	c.823G>A	p.A275T	7	1	6	23	9.15744E-05	T(0.57)	N(-0.17)	B(0.002)	0.002	0.014	
chr12	*SLC39A5*	56629374	c.835G>A	p.G279R	1	0	1	43	0.000152145	T(0.423)	N(-0.48)	P(0.78)	18.66	0.2	
chr12	*SLC39A5*	56629394	c.858_904dupCCCG(	p.E302Afs*117	1	0	1	3	1.19402E-05	NA	NA	NA	NA	NA	
chr12	*SLC39A5*	56629399	c.860C>T	p.P287L	22	0	22	57	0.000201713	D(0.047)	N(-0.57)	B(0.034)	20.3	0.191	Feng CY, et al.

chr12	*SLC39A5*	56629414	c.875T>C	p.L292P	2	0	2	3	1.19384E-05	D(0)	D(-5.11)	D(0.999)	24.6	0.55	
chr12	*SLC39A5*	56629416	c.877G>A	p.G293R	1	0	1	3	1.19384E-05	D(0.001)	D(-3.83)	D(0.993)	25.5	0.408	Wang L, et al.
chr12	*SLC39A5*	56629417	c.878G>C	p.G293A	1	0	1	64	0.000226374	T(0.086)	N(-1.46)	P(0.736)	24	0.12	
chr12	*SLC39A5*	56629471	c.932G>A	p.R311Q	1	0	1	16	5.66436E-05	T(0.249)	N(-0.49)	B(0.006)	16.69	0.034	
chr12	*SLC39A5*	56629474	c.935G>C	p.G312A	1	0	1	/	/	D(0.017)	N(-0.62)	P(0.668)	23.3	0.119	
chr12	*SLC39A5*	56629483	c.944C>A	p.P315Q	1	0	1	/	/	T(0.445)	N(0.81)	P(0.701)	24.2	0.136	
chr12	*SLC39A5*	56630192	c.958C>T	p.R320*	2	0	2	3	1.19307E-05	NA	NA	NA	36	NA	
chr12	*SLC39A5*	56630198	c.964C>T	p.R322*	5	0	5	128	0.000452505	NA	NA	NA	33	NA	
chr12	*SLC39A5*	56630199	c.965G>A	p.R322Q	1	0	1	2	7.95349E-06	T(0.403)	N(0.5)	P(0.763)	18.25	0.116	
chr12	*SLC39A5*	56630203	c.971dupA	p.N324Lfs*59	2	0	1	2	7.95323E-06	NA	NA	NA	NA	NA	
chr12	*SLC39A5*	56630216	c.982C>T	p.R328C	1	0	1	24	8.48404E-05	D(0.031)	N(-0.7)	B(0.376)	16.62	0.099	
chr12	*SLC39A5*	56630217	c.983G>A	p.R328H	1	0	1	70	0.000247453	T(0.289)	N(1.97)	B(0.286)	14.23	0.14	
chr12	*SLC39A5*	56630229	c.995C>T	p.P332L	1	0	1	4	1.59065E-05	T(0.346)	N(-0.41)	B(0.011)	17.77	0.03	
chr12	*SLC39A5*	56630255	c.1022dupA	p.Q341fs	1	0	1	/	/	NA	NA	NA	NA	NA	Zheng YH, et al.
chr12	*SLC39A5*	56630271	c.1037C>T	p.A346V	1	0	1	32	0.000113137	T(0.149)	N(-0.27)	B(0.003)	20.8	0.032	
chr12	*SLC39A5*	56630368	c.1045C>A	p.P349T	2	0	2	1	3.1837E-05	T(0.388)	N(0.57)	B(0.022)	15.2	0.056	
chr12	*SLC39A5*	56630369	c.1046C>A	p.P349Q	3	1	2	5	1.77486E-05	T(0.329)	N(-0.01)	P(0.712)	17.14	0.11	
chr12	*SLC39A5*	56630377	c.1054C>T	p.Q352*	1	0	1	/	/	NA	NA	NA	38	NA	
chr12	*SLC39A5*	56630389	c.1071_1073delGAA	p.K357del	1	0	1	24	8.51619E-05	NA	NA	NA	NA	NA	
chr12	*SLC39A5*	56630394	c.1071G>T	p.K357N	1	0	1	1	3.99278E-06	T(0.214)	N(-0.07)	B(0.016)	13.2	0.036	
chr12	*SLC39A5*	56630404	c.1081C>T	p.H361Y	1	0	1	/	/	T(0.09)	N(-0.81)	B(0.194)	0.188	0.163	
chr12	*SLC39A5*	56630474	c.1151C>T	p.T384M	1	0	1	10	3.55023E-05	D(0.003)	N(-1.25)	P(0.636)	23.1	0.203	
chr12	*SLC39A5*	56630481	c.1158G>T	p.M386I	1	0	1	1	4.01313E-06	D(0.004)	D(-3.65)	D(0.992)	26.8	0.427	
chr12	*SLC39A5*	56630527	c.1204A>G	p.I402V	3	0	3	1	4.19794E-06	D(0.007)	N(-0.65)	P(0.506)	24.2	0.136	

chr12	*SLC39A5*	56630729	c.1222G>A	p.D408N	1	1	0	/	/	T(0.069)	N(-0.26)	P(0.673)	24.1	0.095	
chr12	*SLC39A5*	56630744	c.1237G>A	p.G413S	1	0	1	5	1.76967E-05	D(0)	D(-5.77)	D(1.0)	28.6	0.73	
chr12	*SLC39A5*	56630745	c.1238G>C	p.G413A	1	1	0	/	/	D(0)	D(-5.77)	D(1.0)	27.5	0.904	
chr12	*SLC39A5*	56630975	c.1330C>T	p.R444W	5	0	5	7	2.80359E-05	D(0.018)	D(-4.81)	P(0.828)	22.4	0.193	
chr12	*SLC39A5*	56631093	c.1448G>A	p.G483E	1	0	1	5	2.04705E-05	D(0)	D(-7.69)	B(0.311)	23.7	0.937	
chr12	*SLC39A5*	56631110	c.1465G>A	p.A489T	1	0	1	/	/	D(0)	D(-3.11)	D(0.96)	28.2	0.426	
chr12	*SLC39A5*	56631388	c.1496G>A	p.R499H	1	1	0	24	9.56244E-05	T(1)	N(2.55)	B(0.061)	20.5	0.075	
chr12	*SLC39A5*	56631394	c.1502C>T	p.P501L	1	1	0	274	0.000970633	D(0.035)	N(-1.68)	P(0.888)	24.1	0.135	
chr12	*SLC39A5*	56631409	c.1517C>T	p.T506M	166	9	157	210	0.000744713	T(0.256)	N(-0.01)	B(0.024)	0.681	0.005	
chr12	*SLC39A5*	56631486	c.1594C>T	p.R532W	1	0	1	2	8.96049E-06	D(0.002)	D(-3.4)	B(0.23)	24.9	0.185	
chr12	*SLC39A5*	56631487	c.1595G>A	p.R532Q	2	0	2	11	4.30616E-05	T(0.54)	N(0.9)	B(0.0)	12.93	0.03	
chr05	*P4HA2*	131531143	c.1402G>A	p.V468I	1	1	0	20	7.07774E-05	D(0.002)	N(-0.97)	D(0.984)	28.7	0.558	
chr05	*P4HA2*	131534013	c.1365+1G>C	splicing	2	1	1	7	2.78392E-05	NA	NA	NA	27.6	NA	
chr05	*P4HA2*	131533917	c.1352_1353del	p.V451Gfs*9	4	0	4	3	1.19316E-05	NA	NA	NA	NA	NA	Guo H, et al.
chr05	*P4HA2*	131533921	c.1349G>C	p.R450P	1	0	1	3	1.19312E-05	D(0.000)	N(-6.55)	D(1.000)	31	0.851	
chr05	*P4HA2*	131533955	c.1315C>T	p.R439*	1	0	1	/	/	NA	NA	NA	47	NA	
chr05	*P4HA2*	131539490	c.1202G>A	p.R401Q	1	0	1	4	1.41456E-05	D(0.001)	D(-3.77)	D(0.975)	33	0.646	
chr05	*P4HA2*	131539788	c.1138C>T	p.R380W	3	0	3	3	1.19297E-05	D(0.000)	D(-7.77)	D(1.000)	24.3	0.654	
chr05	*P4HA2*	131539826	c.1100G>A	p.R367H	2	1	1	2	7.07014E-06	T(0.649)	N(0.02)	B(0.009)	23.5	0.139	
chr05	*P4HA2*	131543448	c.1033A>G	p.M345V	1	0	1	1	3.98035E-06	T(0.117)	N(0.15)	B(0.002)	20.2	0.234	
chr05	*P4HA2*	131543457	c.1024T>G	p.Y342D	3	1	2	2	7.96052E-06	D(0.005)	D(-3.29)	B(0.171)	25.6	0.294	
chr05	*P4HA2*	131543489	c.992A>G	p.D331G	1	0	1	/	/	D(0.033)	D(-4.16)	P(0.781)	31	0.575	
chr05	*P4HA2*	131543519	c.962A>G	p.Q321R	1	1	0	/	/	T(0.636)	N(0.27)	B(0.0)	22	0.146	
chr05	*P4HA2*	131543538	c.943C>G	p.H315D	3	0	3	1	4.1953E-06	T(1.0)	N(1.95)	B(0.0)	16.4	0.239	

chr05	*P4HA2*	131543571	c.910C>T	p.R304C	10	0	10	32	0.000134767	D(0.001)	D(-4.65)	P(0.556)	26.6	0.299	
chr05	*P4HA2*	131544847	c.887G>A	p.G296E	1	0	1	/	/	D(0.002)	D(-6.75)	D(0.997)	31	0.578	
chr05	*P4HA2*	131544851	c.883C>T	p.R295C	1	0	1	8	2.83358E-05	D(0.000)	D(-7.83)	D(0.985)	32	0.472	
chr05	*P4HA2*	131544863	c.871G>A	p.E291K	9	2	7	14	5.57387E-05	D(0.001)	D(-3.8)	D(0.984)	32	0.824	Guo H, et al.
chr05	*P4HA2*	131544998	c.736A>T	p.N246Y	1	0	1	1	3.98191E-06	D(0.000)	D(-7.56)	D(0.996)	29.5	0.728	
chr05	*P4HA2*	131545007	c.727G>A	p.A243T	2	0	2	/	/	D(0.001)	D(-3.61)	P(0.806)	29.9	0.473	
chr05	*P4HA2*	131545013	c.721G>A	p.E241K	1	0	1	1	3.98661E-06	T(0.063)	N(-1.75)	B(0.069)	23.6	0.147	
chr05	*P4HA2*	131545988	c.698T>G	p.L233R	1	0	1	/	/	D(0.000)	D(-5.74)	D(0.924)	26.1	0.779	
chr05	*P4HA2*	131545994	c.692delG	p.R231Pfs*32	1	0	1	/	/	NA	NA	NA	NA	NA	
chr05	*P4HA2*	131546009	c.677C>T	p.A226V	1	0	1	1	3.98057E-06	D(0.003)	D(-3.62)	D(0.912)	23.9	0.607	
chr05	*P4HA2*	131546060	c.626T>G	p.V209G	1	0	1	/	/	D(0.000)	D(-6.37)	B(0.342)	23.8	0.362	
chr05	*P4HA2*	131546132	c.554C>T	p.T185M	1	0	1	6	2.12116E-05	D(0.000)	D(-5.2)	D(0.931)	26.8	0.571	
chr05	*P4HA2*	131549659	c.419A>G	p.Q140R	8	0	8	17	6.01047E-05	D(0.001)	D(-3.88)	D(0.993)	27	0.934	Guo H, et al.
chr05	*P4HA2*	131549683	c.395C>A	p.A132D	1	0	1	/	/	D(0.000)	D(-5.69)	D(0.998)	29	0.786	
chr05	*P4HA2*	131549719	c.359G>A	p.R120Q	3	0	3	3	1.19357E-05	D(0.029)	D(-2.94)	P(0.844)	31	0.331	
chr05	*P4HA2*	131552925	c.296C>T	p.A99V	1	0	1	6	2.38698E-05	T(0.11)	N(-0.83)	B(0.105)	22.5	0.15	
chr19	*BSG*	572668	c.34G>A	p.A12T	1	1	0	/	/	T(0.65)	N(-0.44)	B(0.001)	9.58	0.104	
chr19	*BSG*	577776	c.70G>A	p.G24S	24	2	22	9	0.000101338	D(0)	D(-3.45)	D(0.996)	23.9	0.324	
chr19	*BSG*	577792	c.86C>T	p.P29L	1	0	1	6	8.52418E-05	D(0.001)	D(-6.11)	D(1)	23.3	0.301	
chr19	*BSG*	577845	c.139G>T	p.V47L	4	0	4	2	1.21886E-05	D(0.006)	N(-1.41)	B(0.243)	17.73	0.044	
chr19	*BSG*	577855	c.149C>T	p.P50L	1	0	1	/	/	D(0)	D(-5.98)	D(1)	23.9	0.666	
chr19	*BSG*	577863	c.157G>A	p.E53K	1	0	1	10	4.35297E-05	D(0)	N(-2.38)	D(0.979)	25	0.37	
chr19	*BSG*	577869	c.163C>T	p.Q55*	1	0	1	/	/	NA	NA	NA	43	NA	
chr19	*BSG*	577911	c.205C>T	p.Q69*	2	0	2	/	/	NA	NA	NA	42	NA	Jin ZB, et al.

chr19	*BSG*	577947	c.241A>G	p.I81V	2	0	2	/	/	T(0.29)	N(-0.48)	D(0.992)	23	0.192	
chr19	*BSG*	578022	c.316T>C	p.Y106H	1	0	1	/	/	D(0.002)	D(-3.21)	D(1)	23.3	0.425	
chr19	*BSG*	578031	c.325C>T	p.R109W	1	0	1	27	9.78374E-05	D(0.001)	D(-4.75)	D(1)	26	0.537	
chr19	*BSG*	578047	c.341C>T	p.P114L	1	0	1	2	8.24076E-06	D(0)	D(-6.44)	D(1)	25	0.269	
chr19	*BSG*	578091	c.385C>T	p.R129C	4	1	3	4	1.76252E-05	D(0.001)	D(-4.63)	D(0.983)	25.7	0.421	
chr19	*BSG*	578104	c.398T>A	p.V133D	3	0	3	1	4.63302E-06	D(0.012)	N(-0.79)	B(0.087)	17.45	0.139	
chr19	*BSG*	578122	c.415+1G>A	splicing	1	1	0	7	3.30613E-05	/	/	/	29	/	Jin ZB, et al.
chr19	*BSG*	579502	c.418G>C	p.G140R	1	0	1	5	2.01055E-05	T(0.066)	D(-2.71)	P(0.806)	15.26	0.056	
chr19	*BSG*	579557	c.473C>T	p.S158F	2	0	2	2	8.00839E-06	T(0.082)	N(-2.24)	B(0.046)	8.711	0.055	
chr19	*BSG*	579571	c.487G>A	p.A163T	1	0	1	5	1.77895E-05	T(0.305)	N(-0.48)	B(0.004)	0.001	0.018	
chr19	*BSG*	579637	c.553G>A	p.G185S	2	1	1	12	4.35202E-05	T(1.0)	N(0.35)	B(0.067)	8.635	0.087	
chr19	*BSG*	579647	c.563C>T	p.T188M	3	0	3	9	3.28194E-05	T(0.555)	N(-1.56)	P(0.856)	11.08	0.081	
chr19	*BSG*	580389	c.583G>C	p.D195H	140	11	129	178	0.000637381	T(0.111)	N(-1.87)	B(0.105)	14.98	0.077	
chr19	*BSG*	580399	c.593G>T	p.W198L	1	0	1	/	/	T(0.199)	N(-1.07)	B(0.023)	9.234	0.068	
chr19	*BSG*	580428	c.622G>A	p.E208K	2	0	2	15	5.36765E-05	T(0.279)	N(-2.06)	B(0.224)	14.56	0.156	
chr19	*BSG*	580436	c.630G>C	p.M210I	1	0	1	/	/	T(0.184)	N(-0.19)	B(0.0)	1.816	0.034	
chr19	*BSG*	580684	c.694A>C	p.I232L	1	0	1	/	/	T(0.785)	N(-0.23)	B(0.038)	12.38	0.114	
chr19	*BSG*	580700	c.710C>T	p.T237M	2	0	2	3	1.06557E-05	T(0.084)	D(-2.72)	P(0.86)	17.58	0.206	
chr19	*BSG*	580730	c.740T>A	p.V247E	1	0	1	2	7.99603E-06	D(0.007)	N(-1.8)	B(0.102)	12.86	0.102	
chr19	*BSG*	580757	c.767A>C	p.Y256S	1	0	1	/	/	T(0.133)	D(-6.56)	P(0.863)	12.99	0.148	
chr19	*BSG*	580762	c.772A>T	p.I258F	1	0	1	/	/	T(0.078)	N(-1.82)	B(0.279)	0.516	0.282	
chr19	*BSG*	580765	c.775A>G	p.T259A	1	0	1	/	/	T(0.701)	N(-1.01)	B(0.001)	0.061	0.084	
chr19	*BSG*	580766	c.776C>T	p.T259I	2	0	2	/	/	T(0.182)	N(-2.04)	B(0.046)	0.018	0.156	
chr19	*BSG*	580771	c.781T>A	p.S261T	1	0	1	/	/	T(0.269)	N(-1.44)	B(0.001)	0.098	0.167	

chr19	*BSG*	580774	c.784G>A	p.E262K	1	0	1	/	/	T(0.776)	N(-0.93)	B(0.03)	9.462	0.069
chr19	*BSG*	580778	c.788A>G	p.D263G	1	0	1	/	/	T(0.414)	N(-2.17)	B(0.105)	9.424	0.115
chr19	*BSG*	580780	c.790A>C	p.K264Q	1	0	1	/	/	T(0.958)	N(1.12)	B(0.0)	0.249	0.086
chr19	*BSG*	580781	c.791A>G	p.K264R	2	0	2	/	/	T(0.516)	N(-0.24)	B(0.012)	21.9	0.235
chr19	*BSG*	580782	c.792G>T	p.K264N	1	0	1	/	/	T(0.38)	N(-1.1)	B(0.003)	27.1	0.137
chr19	*BSG*	580783	c.792+1G>T	splicing	1	0	1	/	/	NA	NA	NA	33	NA
chr19	*BSG*	581333	c.811G>A	p.E271K	4	0	4	1	4.02557E-06	T(0.163)	N(-1.45)	B(0.009)	9.808	0.172
chr19	*BSG*	581369	c.847T>A	p.S283T	1	0	1	/	/	T(1.0)	N(0.97)	B(0.017)	1.203	0.115
chr19	*BSG*	581405	c.883G>A	p.D295N	1	0	1	/	/	D(0.001)	D(-4.78)	D(0.999)	25.6	0.78
chr19	*BSG*	581418	c.896A>G	p.Y299C	1	0	1	/	/	D(0.0)	D(-8.6)	D(1.0)	23.3	0.762
chr19	*BSG*	581495	c.973C>T	p.L325F	1	0	1	/	/	D(0.029)	D(-3.53)	P(0.669)	24.8	0.194
chr19	*BSG*	581516	c.994G>A	p.V332M	2	0	2	/	/	D(0.005)	D(-2.92)	D(0.992)	29.4	0.287
chr19	*BSG*	582308	c.1072_1074del	p.358_358del	1	0	1	/	/	NA	NA	NA	NA	NA
chr19	*BSG*	582317	c.1081G>A	p.G361S	1	0	1	5	1.87418E-05	D(0.017)	D(-5.26)	P(0.676)	25	0.179
chr19	*BSG*	582535	c.1116G>C	p.Q372H	1	0	1	/	/	NA	NA	NA	18.51	0.037
chr19	*BSG*	582539	c.1120G>A	p.D374N	1	0	1	/	/	T(0.181)	N(-0.56)	B(0.232)	23.5	0.103
chr13	*DZIP1*	96237112	c.2402T>G	p.I801R	1	0	1	/	/	T(0.518)	N(0.91)	B(0.002)	7.901	0.023
chr13	*DZIP1*	96237143	c.2314G>T	p.D772Y	1	0	1	/	/	D(0.001)	D(-6.2)	D(0.993)	26	0.332
chr13	*DZIP1*	96237152	c.2362G>A	p.E788K	1	0	1	/	/	D(0.007)	N(-2.43)	B(0.155)	23	0.055
chr13	*DZIP1*	96237163	c.2351G>T	p.C784F	1	0	1	3	1.22498E-05	T(0.696)	D(-2.6)	B(0.003)	14.43	0.081
chr13	*DZIP1*	96238267	c.2342A>C	p.E781A	1	1	0	1	4.21596E-06	T(0.171)	D(-2.68)	P(0.525)	24.4	0.068
chr13	*DZIP1*	96238373	c.2236C>T	p.P746S	1	1	0	6	2.43942E-05	D(0.014)	N(-0.11)	B(0.044)	13.43	0.033
chr13	*DZIP1*	96238382	c.2227G>A	p.V743I	1	0	1	7	2.55437E-05	T(0.832)	N(0.0)	B(0.002)	0.14	0.023
chr13	*DZIP1*	96238385	c.2224G>A	p.A742T	1	0	1	7	2.898E-05	T(0.776)	N(0.21)	B(0.005)	1.731	0.004

chr13	*DZIP1*	96239858	c.2153A>G	p.K718R	1	0	1	5	1.9885E-05	T(0.37)	N(-1.06)	B(0.036)	12.62	0.069	
chr13	*DZIP1*	96239873	c.2138G>C	p.G713A	67	3	64	85	0.000300525	T(0.318)	N(-1.39)	B(0.03)	0.076	0.023	Lee JK, et al.
chr13	*DZIP1*	96239900	c.2111C>T	p.P704L	1	0	1	4	1.59084E-05	T(0.235)	N(-1.25)	B(0.006)	3.427	0.01	
chr13	*DZIP1*	96239909	c.2102T>A	p.L701H	1	0	1	/	/	D(0.005)	N(-2.28)	D(0.934)	15.81	0.099	
chr13	*DZIP1*	96239940	c.2070_2071insGAC	p.L691delinsDL	2	0	2	/	/	NA	NA	NA	NA	NA	
chr13	*DZIP1*	96241449	c.1983_1985del	p.K661del	3	1	2	4	1.45369E-05	NA	NA	NA	NA	NA	
chr13	*DZIP1*	96242562	c.1814A>C	p.K605T	4	0	4	9	3.18857E-05	D(0.005)	D(-3.49)	B(0.107)	19.57	0.096	
chr13	*DZIP1*	96242599	c.1720C>T	p.Q574*	1	0	1	/	/	NA	NA	NA	NA	NA	
chr13	*DZIP1*	96242685	c.1634G>A	p.G545D	1	0	1	25	9.28436E-05	D(0.0)	D(-5.29)	P(0.787)	23.4	0.166	
chr13	*DZIP1*	96242689	c.1687C>T	p.R563C	1	0	1	5	2.09865E-05	D(0.002)	D(-4.02)	P(0.681)	23.4	0.249	
chr13	*DZIP1*	96251636	c.1520T>G	p.L507R	31	2	29	59	0.000208674	T(0.082)	D(-2.53)	B(0.221)	23	0.117	
chr13	*DZIP1*	96251642	c.1512_1514del	p.E506del	1	0	1	/	/	NA	NA	NA	NA	NA	
chr13	*DZIP1*	96251645	c.1511T>C	p.I504T	1	1	0	5	1.98923E-05	T(0.3)	N(-1.66)	B(0.018)	27.4	0.122	
chr13	*DZIP1*	96251663	c.1493C>T	p.S498L	1	0	1	2	7.95855E-06	T(0.125)	N(-1.34)	B(0.011)	23.5	0.032	
chr13	*DZIP1*	96251672	c.1484C>G	p.A495G	1	1	0	/	/	T(0.188)	N(-1.19)	B(0.015)	21.8	0.024	
chr13	*DZIP1*	96258266	c.1468A>G	p.M490V	1	0	1	1	3.97763E-06	T(0.694)	N(-0.51)	B(0.0)	0.006	0.043	
chr13	*DZIP1*	96261723	c.1408C>T	p.P470S	1	0	1	/	/	D(0.036)	N(-0.73)	B(0.116)	15.74	0.048	
chr13	*DZIP1*	96263705	c.1357C>T	p.P453S	1	0	1	/	/	T(0.424)	N(-1.61)	B(0.001)	10.95	0.037	
chr13	*DZIP1*	96264395	c.1210A>G	p.I404V	3	1	2	/	/	T(0.637)	N(-0.17)	B(0.078)	16.69	0.065	
chr13	*DZIP1*	96264398	c.1207A>G	p.M403V	1	0	1	1	4.02998E-06	T(0.504)	N(-1.2)	B(0.0)	12.18	0.042	
chr13	*DZIP1*	96272137	c.1173+2T>A	splicing	2	0	2	6	2.12355E-05	NA	NA	NA	33	NA	
chr13	*DZIP1*	96272141	c.1171C>T	p.R391W	1	1	0	5	1.99102E-05	D(0.018)	D(-2.62)	P(0.804)	32	0.151	
chr13	*DZIP1*	96274615	c.1092G>A	p.M364I	1	1	0	/	/	T(0.335)	N(-1.24)	B(0.001)	18.24	0.058	
chr13	*DZIP1*	96274646	c.1061C>T	p.S354F	1	0	1	/	/	T(0.074)	N(-1.3)	B(0.103)	20.3	0.038	

chr13	*DZIP1*	96274665	c.1042G>A	p.E348K	7	4	3	14	4.95137E-05	D(0.018)	N(-2.1)	P(0.831)	26.2	0.209
chr13	*DZIP1*	96274670	c.1037C>T	p.A346V	2	0	2	4	1.59146E-05	T(0.708)	N(-0.99)	B(0.022)	16.15	0.026
chr13	*DZIP1*	96274702	c.1005G>C	p.Q335H	5	0	5	13	4.59708E-05	D(0.014)	D(-2.64)	B(0.3)	20.2	0.073
chr13	*DZIP1*	96274706	c.1001T>C	p.M334T	1	0	1	/	/	T(0.278)	N(-1.21)	B(0.288)	21.8	0.118
chr13	*DZIP1*	96277044	c.950C>T	p.S317L	6	1	5	47	0.000174648	D(0.036)	D(-3.75)	B(0.217)	24	0.071
chr13	*DZIP1*	96277076	c.916_918del	p.306_306del	1	0	1	/	/	NA	NA	NA	NA	NA
chr13	*DZIP1*	96277078	c.916G>A	p.E306K	1	1	0	1	4.04855E-06	T(0.329)	N(-1.34)	B(0.272)	24.3	0.077
chr13	*DZIP1*	96277130	c.864G>T	p.R288S	1	0	1	1	4.01101E-06	T(0.262)	D(-2.84)	B(0.044)	13.54	0.063
chr13	*DZIP1*	96277151	c.843C>G	p.D281E	2	1	1	6	2.15558E-05	T(1.0)	N(0.62)	B(0.004)	9.541	0.034
chr13	*DZIP1*	96282260	c.793G>A	p.A265T	1	0	1	6	2.15558E-05	T(0.337)	N(-0.51)	B(0.239)	13.75	0.061
chr13	*DZIP1*	96282302	c.751C>A	p.Q251K	1	1	0	1	3.98067E-06	T(0.452)	N(-0.74)	D(0.997)	23.8	0.154
chr13	*DZIP1*	96282320	c.733G>C	p.V245L	2	0	2	3	1.19464E-05	T(0.383)	N(-0.26)	B(0.022)	11.55	0.031
chr13	*DZIP1*	96282334	c.719G>A	p.R240Q	1	1	0	/	/	T(1.0)	N(1.53)	B(0.001)	14.77	0.077
chr13	*DZIP1*	96282335	c.718C>T	p.R240W	1	0	1	7	2.79272E-05	D(0.016)	D(-2.97)	B(0.003)	23.9	0.13
chr13	*DZIP1*	96293581	c.565C>A	p.L189M	1	0	1	/	/	T(0.324)	N(-0.61)	B(0.074)	13.78	0.011
chr13	*DZIP1*	96293593	c.553T>C	p.S185P	1	0	1	/	/	D(0.011)	D(-3.0)	P(0.806)	20.8	0.093
chr13	*DZIP1*	96293658	c.488T>C	p.L163P	2	0	2	/	/	D(0.001)	D(-5.09)	D(0.962)	20.7	0.189
chr13	*DZIP1*	96293724	c.422A>G	p.Q141R	1	0	1	1	3.98565E-06	T(0.053)	N(-1.97)	D(0.989)	22.4	0.14
chr13	*DZIP1*	96293760	c.386A>G	p.E129G	1	0	1	3	1.0629E-05	D(0.0)	D(-6.44)	D(0.999)	27.5	0.634
chr13	*DZIP1*	96293764	c.382A>G	p.I128V	1	0	1	/	/	T(0.196)	N(-0.56)	P(0.644)	22.1	0.1
chr13	*DZIP1*	96293822	c.324C>A	p.H108Q	1	0	1	/	/	D(0.021)	D(-3.77)	D(0.993)	20.3	0.186
chr13	*DZIP1*	96293953	c.193G>A	p.E65K	1	0	1	/	/	D(0.005)	D(-3.41)	D(0.999)	25.7	0.347
chr13	*DZIP1*	96293958	c.188G>A	p.R63Q	1	0	1	/	/	D(0.0)	D(-3.68)	D(0.999)	25.8	0.408
chr13	*DZIP1*	96294052	c.94G>T	p.A32S	1	0	1	/	/	T(1.0)	N(0.26)	B(0.001)	0.001	0.033

chr13	*DZIP1*	96294096	c.50A>T	p.H17L	1	0	1	3	9.74089E-05	D(0.003)	D(-3.23)	P(0.466)	22.9	0.074
chr16	*XYLT1*	17202647	c.2785G>T	p.V929F	1	0	1	/	/	D(0.045)	N(-1.87)	D(0.972)	23.5	0.207
chr16	*XYLT1*	17202649	c.2783C>T	p.P928L	1	0	1	6	2.14359E-05	T(0.117)	D(-5.65)	D(0.934)	22.9	0.345
chr16	*XYLT1*	17202693	c.2739G>A	p.M913I	9	0	9	7	2.48777E-05	T(0.1)	N(-0.5)	B(0.001)	16.42	0.055
chr16	*XYLT1*	17202704	c.2728delG	p.V910Wfs*74	1	0	1	/	/	NA	NA	NA	NA	
chr16	*XYLT1*	17202733	c.2699C>T	p.T900I	1	0	1	1	3.99632E-06	T(0.151)	N(-1.53)	B(0.035)	10.44	0.039
chr16	*XYLT1*	17202742	c.2690C>T	p.S897F	5	2	3	6	2.39754E-05	T(0.297)	N(-0.03)	B(0.002)	12.98	0.055
chr16	*XYLT1*	17202743	c.2689T>C	p.S897P	1	0	1	/	/	T(0.114)	N(-1.45)	B(0.169)	12.98	0.072
chr16	*XYLT1*	17202835	c.2597C>T	p.A866V	1	0	1	/	/	T(0.249)	N(-1.01)	P(0.585)	23.3	0.141
chr16	*XYLT1*	17202842	c.2590C>T	p.R864C	1	0	1	6	2.78458E-05	D(0.032)	D(-3.16)	P(0.855)	25.4	0.193
chr16	*XYLT1*	17202853	c.2579A>G	p.N860S	1	0	1	1	4.97676E-06	T(0.331)	N(-0.24)	B(0.007)	7.411	0.138
chr16	*XYLT1*	17202871	c.2561A>C	p.E854A	22	0	22	67	0.000312599	T(0.149)	D(-3.08)	B(0.256)	24.2	0.129
chr16	*XYLT1*	17211608	c.2452C>T	p.P818S	1	0	1	/	/	D(0.007)	D(-7.32)	D(0.999)	27.7	0.643
chr16	*XYLT1*	17211656	c.2404A>G	p.T802A	1	0	1	/	/	T(0.141)	N(-0.93)	B(0.073)	22.2	0.127
chr16	*XYLT1*	17211674	c.2386G>A	p.D796N	1	0	1	1	3.97636E-06	D(0.047)	D(-4.05)	D(0.998)	29.2	0.363
chr16	*XYLT1*	17211682	c.2378C>T	p.A793V	2	0	2	4	1.59057E-05	D(0.031)	N(-2.43)	D(1)	28.3	0.541
chr16	*XYLT1*	17211686	c.2374G>A	p.A792T	1	0	1	4	1.59056E-05	T(0.068)	D(-3.01)	B(0.387)	24.7	0.325
chr16	*XYLT1*	17211722	c.2338G>A	p.V780M	1	0	1	34	0.000120202	D(0.007)	N(-1.79)	D(0.995)	27.3	0.235
chr16	*XYLT1*	17211748	c.2312A>C	p.K771T	1	1	0	1	3.97671E-06	T(0.067)	D(-3.54)	D(0.971)	24.3	0.212
chr16	*XYLT1*	17211799	c.2261G>A	p.R754H	2	1	1	18	6.38257E-05	D(0.001)	D(-4.73)	D(1)	31	0.738
chr16	*XYLT1*	17211833	c.2227G>A	p.G743S	2	1	1	4	1.61998E-05	T(0.225)	D(-4.45)	D(1)	25.1	0.361
chr16	*XYLT1*	17228367	c.1990G>A	p.E664K	3	0	3	9	3.18356E-05	T(0.755)	N(-0.44)	P(0.464)	23.4	0.173
chr16	*XYLT1*	17228372	c.1985G>A	p.R662Q	8	3	5	34	0.000120273	T(0.272)	N(-1.06)	B(0.257)	23.7	0.174
chr16	*XYLT1*	17228381	c.1976G>A	p.G659D	2	0	2	/	/	D(0.014)	D(-3.58)	D(1)	28.8	0.655

chr16	*XYLT1*	17228421	c.1936G>A	p.D646N	1	0	1	/	/	D(0)	D(-4.47)	D(0.967)	29.6	0.37	
chr16	*XYLT1*	17228439	c.1918G>A	p.G640S	6	2	4	154	0.000544447	T(0.203)	D(-2.87)	P(0.892)	25.1	0.236	
chr16	*XYLT1*	17228474	c.1883G>A	p.R628H	1	0	1	8	2.82833E-05	T(0.538)	N(-1.05)	B(0.055)	23	0.058	
chr16	*XYLT1*	17228483	c.1874C>T	p.P625L	1	0	1	27	9.54549E-05	D(0.035)	D(-5.8)	P(0.906)	24.7	0.283	
chr16	*XYLT1*	17228516	c.1829_1841del	p.G610Vfs*31	1	1	0	/	/	NA	NA	NA	NA		
chr16	*XYLT1*	17228520	c.1837G>T	p.D613Y	2	0	2	/	/	D(0)	D(-7.28)	D(0.991)	31	0.644	
chr16	*XYLT1*	17228550	c.1807G>A	p.V603M	1	0	1	57	0.000201546	D(0.034)	N(-1.06)	P(0.488)	23.3	0.051	
chr16	*XYLT1*	17228583	c.1774C>T	p.R592W	2	0	2	3	1.19386E-05	D(0)	D(-6.75)	D(0.969)	29.5	0.346	
chr16	*XYLT1*	17232210	c.1764+2T>C	splicing	1	0	1	/	/	NA	NA	NA	32	#NNAA	
chr16	*XYLT1*	17232219	c.1757G>A	p.R586H	1	0	1	5	1.99173E-05	D(0.001)	D(-4.69)	D(0.994)	31	0.525	
chr16	*XYLT1*	17232220	c.1756C>T	p.R586C	1	0	1	63	0.000223026	D(0)	D(-7.41)	D(0.994)	31	0.315	
chr16	*XYLT1*	17232259	c.1717G>A	p.G573S	1	0	1	23	8.13209E-05	D(0)	D(-5.8)	D(1)	27.9	0.578	
chr16	*XYLT1*	17232271	c.1705G>A	p.V569M	1	0	1	4	1.59066E-05	D(0)	D(-2.9)	D(0.984)	27.6	0.46	
chr16	*XYLT1*	17232306	c.1670G>A	p.R557H	1	0	1	4	1.59056E-05	D(0.001)	D(-4.83)	D(0.984)	31	0.333	
chr16	*XYLT1*	17232324	c.1652G>A	p.R551H	1	0	1	9	3.57878E-05	T(0.319)	N(-2.27)	D(0.97)	26.4	0.445	
chr16	*XYLT1*	17232353	c.1623C>G	p.H541Q	1	0	1	/	/	T(0.236)	N(-2.16)	P(0.876)	22.7	0.212	
chr16	*XYLT1*	17232375	c.1601C>T	p.T534M	3	1	2	16	6.36826E-05	D(0)	D(-5.83)	D(0.984)	28.8	0.716	
chr16	*XYLT1*	17235071	c.1523_1525delTCT	p.F508del	2	0	2	2	7.95311E-06	NA	NA	NA	NA		
chr16	*XYLT1*	17235132	c.1465G>A	p.V489M	1	0	1	9	3.18246E-05	T(0.064)	N(-1.9)	D(0.998)	25.2	0.246	Lee JK, et al.
chr16	*XYLT1*	17235180	c.1417G>A	p.A473T	1	0	1	/	/	T(1)	N(0.87)	B(0.011)	17.92	0.08	
chr16	*XYLT1*	17235207	c.1390C>G	p.L464V	1	1	0	/	/	D(0)	D(-2.9)	D(0.999)	27	0.309	
chr16	*XYLT1*	17252703	c.1353C>A	p.H451Q	1	0	1	/	/	D(0.001)	D(-7.69)	D(1)	22.6	0.397	
chr16	*XYLT1*	17252728	c.1328G>A	p.R443Q	2	0	2	13	4.59611E-05	D(0.015)	D(-3.75)	D(0.999)	29.6	0.304	
chr16	*XYLT1*	17252746	c.1310C>T	p.A437V	1	1	0	16	5.65723E-05	D(0.043)	D(-2.64)	D(0.985)	28.2	0.23	

chr16	*XYLT1*	17252749	c.1307T>C	p.V436A	1	0	1	11	4.37459E-05	D(0.009)	D(-3.39)	D(0.997)	27.2	0.362	
chr16	*XYLT1*	17292087	c.1271C>T	p.A424V	1	1	0	4	1.42148E-05	D(0.017)	D(-2.99)	D(0.993)	29.7	0.545	
chr16	*XYLT1*	17292121	c.1237G>A	p.D413N	1	0	1	15	5.31026E-05	T(0.196)	D(-2.37)	D(1)	27.1	0.245	
chr16	*XYLT1*	17292129	c.1229A>T	p.E410V	1	0	1	4	1.59258E-05	D(0.026)	D(-4.35)	P(0.577)	27.2	0.176	
chr16	*XYLT1*	17292142	c.1216C>T	p.R406W	2	1	1	20	7.07704E-05	D(0.002)	D(-4.64)	D(0.957)	25.6	0.293	
chr16	*XYLT1*	17292182	c.1176G>A	p.W392*	1	0	1	/	/	NA	NA	NA	43	#NNAA	
chr16	*XYLT1*	17292252	c.1106G>A	p.R369Q	1	0	1	3	1.19715E-05	D(0.005)	D(-3.81)	D(0.999)	32	0.365	
chr16	*XYLT1*	17294392	c.1033A>C	p.M345L	1	0	1	37	0.000130822	T(1)	N(1.1)	B(0.018)	17.74	0.122	
chr16	*XYLT1*	17294449	c.976G>T	p.V326F	1	0	1	/	/	D(0.001)	D(-4.35)	D(0.972)	28.2	0.481	
chr16	*XYLT1*	17352862	c.896G>A	p.R299Q	4	1	3	6	2.89226E-05	T(0.101)	D(-2.6)	D(0.996)	24.8	0.333	Lee JK, et al.
chr16	*XYLT1*	17352901	c.857G>A	p.R286H	1	0	1	1	5.56347E-06	T(0.102)	N(-1.84)	P(0.584)	23.7	0.11	
chr16	*XYLT1*	17352919	c.839T>C	p.I280T	1	0	1	2	9.30224E-06	D(0)	D(-4.93)	D(0.993)	26.7	0.662	
chr16	*XYLT1*	17352928	c.830G>A	p.R277H	1	0	1	5	2.28795E-05	D(0.003)	D(-3.29)	D(0.997)	29.6	0.341	
chr16	*XYLT1*	17353002	c.756C>A	p.D252E	1	0	1	/	/	T(0.321)	N(-0.34)	B(0.004)	19.54	0.104	
chr16	*XYLT1*	17353034	c.724A>G	p.T242A	3	0	3	7	2.56694E-05	T(0.316)	N(-0.15)	B(0.001)	6.661	0.015	
chr16	*XYLT1*	17353066	c.692A>G	p.K231R	3	0	3	4	1.59252E-05	T(0.318)	N(-0.27)	B(0.056)	19.59	0.13	
chr16	*XYLT1*	17353118	c.640G>A	p.G214S	2	1	1	5	1.98938E-05	T(0.242)	N(0.28)	B(0)	10.77	0.041	
chr16	*XYLT1*	17353201	c.557C>T	p.P186L	3	2	1	6	2.12116E-05	T(0.427)	N(-0.05)	B(0.001)	13.84	0.068	
chr16	*XYLT1*	17353318	c.440G>T	p.R147L	1	0	1	/	/	D(0.011)	N(-1.5)	P(0.652)	25.4	0.092	
chr16	*XYLT1*	17353333	c.425C>T	p.P142L	1	0	1	35	0.000124658	D(0)	D(-2.76)	D(0.998)	27	0.236	
chr16	*XYLT1*	17353340	c.418C>T	p.H140Y	1	0	1	/	/	D(0.002)	N(-1.68)	D(0.99)	27	0.229	
chr16	*XYLT1*	17353351	c.407G>A	p.G136D	1	0	1	/	/	D(0.024)	N(-1.73)	P(0.623)	24.7	0.107	
chr16	*XYLT1*	17451882	c.389C>T	p.T130I	3	0	3	11	3.89819E-05	T(0.051)	N(-0.33)	B(0.076)	20.4	0.031	
chr16	*XYLT1*	17451891	c.380C>T	p.P127L	1	0	1	7	2.79318E-05	D(0.018)	N(0.08)	D(0.997)	24	0.149	

chr16	*XYLT1*	17564563	c.91G>A	p.V31I	1	0	1	1	4.00994E-05	T(1)	N(-0.06)	B(0)	11.11	0.031	
chr02	*NDUFAF7*	37458854	c.6T>G	p.S2R	1	1	0	2	7.07564E-06	D(0.024)	N(0.33)	B(0.001)	0.846	0.029	
chr02	*NDUFAF7*	37459249	c.56C>T	p.A19V	1	1	0	10	3.97678E-05	T(0.296)	N(0.08)	B(0.0)	10.87	0.023	
chr02	*NDUFAF7*	37459288	c.95G>T	p.G32V	2	0	2	/	/	D(0.011)	N(-0.69)	P(0.731)	21.9	0.086	
chr02	*NDUFAF7*	37459344	c.151A>G	p.I51V	1	0	1	3	1.06045E-05	D(0.02)	N(-0.98)	B(0.32)	23.2	0.458	
chr02	*NDUFAF7*	37459396	c.203C>G	p.T68S	1	0	1	2	7.95425E-06	D(0.007)	D(-3.57)	P(0.589)	24.8	0.599	
chr02	*NDUFAF7*	37463255	c.233G>T	p.R78L	4	1	3	39	0.000138114	D(0.011)	D(-4.24)	B(0.118)	19.74	0.375	
chr02	*NDUFAF7*	37468815	c.503C>T	p.T168I	1	0	1	8	2.82963E-05	T(0.236)	N(-2.1)	P(0.496)	19.18	0.338	
chr02	*NDUFAF7*	37468866	c.554A>T	p.Y185F	1	0	1	/	/	D(0.006)	D(-3.41)	D(0.992)	23.6	0.292	
chr02	*NDUFAF7*	37468869	c.557T>C	p.M186T	1	0	1	/	/	T(0.141)	N(-1.02)	B(0.443)	22.2	0.198	
chr02	*NDUFAF7*	37468893	c.581T>A	p.I194N	2	0	2	6	2.45138E-05	D(0.001)	D(-4.15)	P(0.797)	27.4	0.578	
chr02	*NDUFAF7*	37468896	c.584C>T	p.P195L	1	1	0	/	/	D(0.009)	D(-6.02)	P(0.566)	27.6	0.701	
chr02	*NDUFAF7*	37469792	c.637C>G	p.L213V	17	0	17	75	0.000265858	T(0.254)	N(-0.92)	B(0.163)	20.3	0.32	
chr02	*NDUFAF7*	37473201	c.799G>A	p.E267K	6	0	6	17	6.77199E-05	T(0.108)	D(-2.53)	B(0.362)	26.1	0.376	
chr02	*NDUFAF7*	37474622	c.960T>A	p.H320Q	1	1	0	3	1.19339E-05	D(0.011)	D(-6.63)	D(0.99)	23	0.503	Liu F, et al.
chr02	*NDUFAF7*	37474765	c.1103G>T	p.R368L	1	0	1	6	2.14272E-05	D(0.0)	D(-6.94)	D(0.999)	31	0.916	
chr02	*NDUFAF7*	37474767	c.1105C>G	p.L369V	1	0	1	1	4.02379E-06	D(0.003)	D(-2.71)	P(0.688)	23	0.56	
chr02	*NDUFAF7*	37475432	c.1265C>T	p.A422V	1	0	1	1	3.97719E-06	D(0.029)	N(-0.29)	B(0.008)	11.38	0.072	
chr02	*NDUFAF7*	37475435	c.1268G>A	p.R423H	1	0	1	13	4.59621E-05	T(0.184)	N(-0.12)	B(0.103)	15.6	0.047	
chr08	*CPSF1*	145618552	c.4313G>A	p.R1438H	5	0	5	2	1.25356E-05	D(0.037)	D(-2.98)	D(0.972)	23.6	0.169	
chr08	*CPSF1*	145618558	c.4307C>T	p.T1436M	2	0	2	5	3.86739E-05	D(0.01)	N(0.72)	B(0.001)	23.7	0.107	
chr08	*CPSF1*	145618726	c.4225T>C	p.Y1409H	1	0	1	/	/	T(0.46)	N(0.29)	B(0.028)	22.4	0.193	
chr08	*CPSF1*	145618789	c.4162C>T	p.R1388C	1	0	1	2	8.61171E-06	T(0.073)	D(-3.51)	D(0.997)	23.1	0.317	
chr08	*CPSF1*	145618803	c.4148T>C	p.M1383T	1	0	1	/	/	T(1)	N(-0.35)	B(0.013)	20.8	0.237	

chr08	*CPSF1*	145618807	c.4146-2A>G	splicing	1	1	0	1	4.34938E-06	/	/	/	34	/	Ouyang JM, et al.
chr08	*CPSF1*	145618883	c.4144C>T	p.R1382W	1	0	1	6	3.42853E-05	D(0)	D(-7.14)	D(1)	33	0.714	
chr08	*CPSF1*	145618894	c.4133C>T	p.P1378L	1	0	1	/	/	D(0)	D(-7.56)	D(0.997)	26.9	0.543	
chr08	*CPSF1*	145618907	c.4120G>A	p.A1374T	27	3	24	18	8.75435E-05	D(0.008)	D(-3.08)	D(1)	24.7	0.397	
chr08	*CPSF1*	145618920	c.4107G>A	p.M1369I	2	0	2	1	5.55142E-06	T(0.337)	N(-1.45)	B(0.08)	19.1	0.118	
chr08	*CPSF1*	145618997	c.4030G>A	p.G1344S	1	0	1	8	3.49433E-05	T(0.954)	N(-0.5)	B(0.213)	18.76	0.196	
chr08	*CPSF1*	145619128	c.3985G>A	p.V1329M	1	0	1	9	3.22283E-05	T(0.265)	N(-0.11)	B(0.002)	15.45	0.022	
chr08	*CPSF1*	145619131	c.3982G>A	p.V1328I	1	0	1	118	0.000421947	T(0.46)	N(-0.15)	B(0.0)	15.88	0.095	
chr08	*CPSF1*	145619133	c.3980C>T	p.S1327L	7	4	3	117	0.000418225	T(0.293)	N(-2.12)	B(0.002)	23.2	0.075	
chr08	*CPSF1*	145619139	c.3974A>G	p.K1325R	1	0	1	/	/	T(0.654)	N(-0.6)	B(0.005)	20	0.061	
chr08	*CPSF1*	145619148	c.3965G>C	p.G1322A	3	0	3	1	4.0151E-06	T(0.679)	N(-0.63)	B(0.0)	17.91	0.069	
chr08	*CPSF1*	145619157	c.3956C>T	p.A1319V	2	0	2	9	3.61803E-05	T(0.615)	N(-1.38)	B(0.008)	20.8	0.115	
chr08	*CPSF1*	145619164	c.3949C>T	p.R1317W	1	0	1	11	4.41044E-05	D(0.007)	D(-4.7)	D(1)	27.4	0.465	
chr08	*CPSF1*	145619251	c.3862_3871dup	p.F1291*	1	1	0	/	/	NA	NA		NA	NA	Ouyang JM, et al.
chr08	*CPSF1*	145619333	c.3854C>T	p.P1285L	1	1	0	2	8.02369E-06	D(0.0)	D(-9.61)	D(1)	28.1	0.609	
chr08	*CPSF1*	145619342	c.3845T>C	p.M1282T	1	1	0	1	4.00921E-06	T(0.109)	D(-3.24)	B(0.274)	22.7	0.208	
chr08	*CPSF1*	145619364	c.3823G>T	p.D1275Y	1	1	0	/	/	D(0.002)	D(-4.39)	D(0.999)	25.2	0.492	Ouyang JM, et al.
chr08	*CPSF1*	145619364	c.3823G>A	p.D1275N	1	1	0	1	4.0151E-06	D(0.008)	N(-1.94)	P(0.784)	23.8	0.096	
chr08	*CPSF1*	145619367	c.3820C>T	p.R1274C	1	1	0	12	4.81928E-05	T(0.066)	D(-2.61)	D(0.998)	27.2	0.25	
chr08	*CPSF1*	145619479	c.3781A>G	p.M1261V	5	0	5	3	1.24259E-05	T(0.22)	N(-1.57)	B(0.003)	21.2	0.116	
chr08	*CPSF1*	145619488	c.3772G>T	p.V1258L	1	0	1	/	/	D(0.032)	N(-1.88)	B(0.218)	22.6	0.123	
chr08	*CPSF1*	145619626	c.3712G>A	p.E1238K	3	0	3	4	1.66287E-05	D(0.02)	D(-3.2)	P(0.731)	26	0.233	
chr08	*CPSF1*	145619674	c.3664A>C	p.I1222L	1	0	1	/	/	T(0.053)	N(-1.39)	B(0.173)	22.7	0.155	
chr08	*CPSF1*	145619696	c.3642G>C	p.Q1214H	1	0	1	1	4.15766E-06	T(0.073)	D(-3.14)	P(0.74)	23.2	0.22	

chr08	*CPSF1*	145619890	c.3536A>G	p.N1179S	1	0	1	1	4.10028E-06	T(0.761)	N(-1.37)	B(0.024)	16.98	0.117	
chr08	*CPSF1*	145619906	c.3520G>A	p.A1174T	3	0	3	10	3.5978E-05	D(0.019)	D(-3.12)	D(0.998)	27.1	0.422	
chr08	*CPSF1*	145620138	c.3373A>T	p.T1125S	2	0	2	/	/	D(0.0)	D(-3.71)	D(1)	26.1	0.493	
chr08	*CPSF1*	145620144	c.3367G>A	p.A1123T	1	1	0	8	0.000028685	D(0.029)	N(-0.64)	P(0.529)	23.1	0.148	
chr08	*CPSF1*	145620147	c.3364G>A	p.A1122T	1	0	1	/	/	D(0.04)	D(-2.72)	D(0.999)	24.5	0.261	
chr08	*CPSF1*	145620150	c.3361G>A	p.V1121M	1	0	1	5	2.01665E-05	D(0.005)	N(-1.16)	P(0.942)	25.6	0.189	
chr08	*CPSF1*	145620171	c.3340G>A	p.V1114M	4	0	4	4	1.43122E-05	T(0.237)	N(0.08)	P(0.725)	23	0.139	
chr08	*CPSF1*	145620176	c.3335A>G	p.E1112G	1	0	1	1	4.0318E-06	T(0.651)	N(1.67)	B(0.004)	22.6	0.204	
chr08	*CPSF1*	145620194	c.3317T>C	p.V1106A	1	1	0	/	/	T(0.094)	D(-3.41)	D(0.998)	25.8	0.505	
chr08	*CPSF1*	145620235	c.3276C>G	p.I1092M	1	0	1	2	7.23223E-06	T(0.124)	N(-1.42)	P(0.493)	17.56	0.1	
chr08	*CPSF1*	145620353	c.3232C>T	p.L1078F	2	0	2	/	/	D(0.003)	D(-3.64)	D(1)	27.5	0.495	
chr08	*CPSF1*	145620359	c.3226A>G	p.I1076V	1	0	1	/	/	T(0.465)	N(-0.53)	B(0.246)	21.3	0.113	
chr08	*CPSF1*	145620542	c.3125C>T	p.T1042M	22	0	22	23	9.19647E-05	D(0.029)	N(-1.19)	B(0.007)	22.3	0.131	
chr08	*CPSF1*	145620545	c.3122A>G	p.N1041S	1	0	1	1	3.9991E-06	T(0.935)	N(0.54)	B(0.0)	13.31	0.137	
chr08	*CPSF1*	145621588	c.2954G>A	p.R985H	6	0	6	34	0.000141333	T(0.164)	N(-0.67)	B(0.001)	14.74	0.269	
chr08	*CPSF1*	145621637	c.2905G>A	p.D969N	1	0	1	/	/	T(0.196)	D(-3.77)	P(0.933)	23.1	0.283	
chr08	*CPSF1*	145621657	c.2885G>A	p.R962Q	1	0	1	7	3.17676E-05	D(0.01)	D(-3.01)	P(0.909)	24.2	0.256	
chr08	*CPSF1*	145621658	c.2884C>T	p.R962W	1	0	1	4	1.80719E-05	D(0.001)	D(-6.7)	D(1)	28.5	0.59	
chr08	*CPSF1*	145621686	c.2856G>T	p.W952C	1	0	1	/	/	D(0.001)	D(-9.33)	D(1)	27.5	0.635	
chr08	*CPSF1*	145621814	c.2823_2824del	p.V943Lfs*65	1	1	0	22	7.83839E-05	NA	NA	NA	NA	NA	Ouyang JM, et al.
chr08	*CPSF1*	145621859	c.2780G>A	p.R927H	7	1	6	14	4.97728E-05	D(0.01)	N(-1.89)	P(0.75)	23.4	0.124	
chr08	*CPSF1*	145621865	c.2774G>A	p.R925Q	1	0	1	7	2.48853E-05	T(0.654)	N(-0.01)	B(0.017)	21.7	0.062	
chr08	*CPSF1*	145621866	c.2773C>G	p.R925G	2	0	2	/	/	T(0.339)	N(-1.77)	B(0.0)	22.7	0.081	
chr08	*CPSF1*	145621869	c.2770G>A	p.A924T	1	1	0	150	0.00053317	T(0.398)	N(0.1)	B(0.0)	11.05	0.025	

chr08	*CPSF1*	145622087	c.2650G>A	p.G884S	2	0	2	8	2.83628E-05	T(0.846)	N(0.85)	B(0.0)	15.39	0.022	
chr08	*CPSF1*	145622434	c.2580delG	p.R860Sfs*96	1	0	1	/	/	NA	NA	NA	NA	NA	
chr08	*CPSF1*	145622445	c.2569C>T	p.R857C	6	1	5	12	6.57239E-05	T(0.078)	D(-3.89)	P(0.927)	22.8	0.18	
chr08	*CPSF1*	145622447	c.2567G>T	p.S856I	1	0	1	/	/	T(0.379)	N(-1.22)	B(0.0)	19.36	0.083	
chr08	*CPSF1*	145622450	c.2564G>T	p.G855V	2	0	2	/	/	D(0)	D(-8.49)	D(1)	24.5	0.639	
chr08	*CPSF1*	145622498	c.2516G>A	p.R839H	1	0	1	2	1.3129E-05	T(0.128)	N(-0.82)	D(0.997)	24.1	0.16	
chr08	*CPSF1*	145622501	c.2513C>T	p.T838M	1	0	1	141	0.000764534	T(0.154)	N(-1.15)	P(0.954)	23	0.082	
chr08	*CPSF1*	145622613	c.2401G>A	p.D801N	1	0	1	2	1.097E-05	D(0.014)	D(-3.92)	D(0.992)	26.6	0.428	
chr08	*CPSF1*	145622722	c.2365C>T	p.R789W	1	0	1	24	9.04146E-05	D(0)	D(-7.33)	D(1)	31	0.378	
chr08	*CPSF1*	145622764	c.2323G>A	p.A775T	1	0	1	/	/	T(0.65)	N(-0.55)	B(0.0)	12.64	0.034	
chr08	*CPSF1*	145622767	c.2320C>T	p.P774S	1	0	1	/	/	T(0.574)	N(-0.73)	B(0.003)	19.78	0.066	
chr08	*CPSF1*	145622772	c.2315G>A	p.R772Q	4	0	4	4	1.64391E-05	T(0.537)	N(-0.42)	B(0.01)	22	0.032	
chr08	*CPSF1*	145622773	c.2314C>T	p.R772W	6	0	6	8	3.78544E-05	T(0.217)	N(-0.79)	D(0.98)	23.3	0.139	
chr08	*CPSF1*	145622795	c.2292A>C	p.R764S	5	0	5	96	0.00041301	T(0.275)	N(-0.81)	B(0.033)	20.6	0.173	Zheng YH, et al.
chr08	*CPSF1*	145622835	c.2252C>T	p.S751L	6	0	6	7	3.59059E-05	D(0.007)	D(-3.61)	P(0.915)	25.4	0.163	
chr08	*CPSF1*	145622998	c.2170C>T	p.R724C	4	0	4	22	8.10498E-05	D(0.046)	N(-2.21)	D(0.977)	23.5	0.117	
chr08	*CPSF1*	145623004	c.2164_2165insGG	p.R724Afs*86	1	0	1	/	/	NA	NA	NA	NA	NA	
chr08	*CPSF1*	145623007	c.2161C>G	p.L721V	1	1	0	7	2.90028E-05	T(0.545)	N(-0.07)	B(0.009)	9.008	0.007	
chr08	*CPSF1*	145623013	c.2155G>C	p.D719H	1	0	1	/	/	D(0.023)	N(-1.92)	P(0.576)	22.8	0.084	
chr08	*CPSF1*	145623015	c.2153G>A	p.R718H	1	0	1	2	8.24328E-06	T(0.093)	N(-0.87)	B(0.001)	22.4	0.073	
chr08	*CPSF1*	145623016	c.2152C>T	p.R718C	1	0	1	6	2.47243E-05	D(0.041)	N(-2.26)	B(0.203)	24.5	0.092	
chr08	*CPSF1*	145623066	c.2102A>T	p.Y701F	1	0	1	/	/	D(0.03)	D(-3.43)	D(0.999)	24.3	0.234	
chr08	*CPSF1*	145623078	c.2090C>T	p.T697M	1	0	1	4	1.61633E-05	D(0.024)	N(-1.97)	P(0.482)	22.7	0.158	
chr08	*CPSF1*	145623087	c.2081A>G	p.K694R	2	0	2	1	4.04655E-06	T(0.64)	N(-0.99)	B(0.001)	18.57	0.09	

chr08	*CPSF1*	145623194	c.2048C>T	p.A683V	1	0	1	1	3.19836E-05	T(0.476)	N(-1.41)	B(0.022)	22.8	0.114	
chr08	*CPSF1*	145623209	c.2033G>A	p.R678H	1	0	1	5	1.80074E-05	T(0.148)	N(-0.84)	B(0.002)	22.9	0.065	
chr08	*CPSF1*	145623210	c.2032C>T	p.R678C	8	1	7	20	7.18943E-05	D(0.022)	N(-1.12)	P(0.857)	23.2	0.125	
chr08	*CPSF1*	145623216	c.2026G>A	p.G676S	1	0	1	5	1.79107E-05	T(0.171)	N(-0.59)	B(0.047)	17.48	0.046	
chr08	*CPSF1*	145623230	c.2012delA	p.K671Rfs*25	1	0	1	/	/	NA	NA	NA	NA	NA	
chr08	*CPSF1*	145623266	c.1976T>C	p.M659T	5	0	5	3	1.20504E-05	D(0.012)	D(-3.16)	P(0.623)	23.9	0.432	
chr08	*CPSF1*	145623278	c.1964A>G	p.Y655C	1	0	1	3	1.20649E-05	D(0.001)	D(-8.57)	D(1.0)	25.9	0.346	
chr08	*CPSF1*	145623312	c.1930G>A	p.A644T	1	0	1	3	1.22508E-05	T(0.091)	N(-0.88)	B(0.007)	18.23	0.057	
chr08	*CPSF1*	145623598	c.1882_1894del	p.L628*	2	0	2	/	/	NA	NA	NA	NA	NA	
chr08	*CPSF1*	145623719	c.1867C>A	p.P623T	1	1	0	/	/	T(0.52)	D(-2.62)	B(0.213)	22	0.231	Ouyang JM, et al.
chr08	*CPSF1*	145623728	c.1858C>T	p.Q620*	1	1	0	/	/	NA	NA	NA	39	NA	
chr08	*CPSF1*	145623746	c.1840G>T	p.D614Y	1	0	1	/	/	D(0.019)	D(-4.68)	D(0.984)	31	0.386	
chr08	*CPSF1*	145623749	c.1837G>A	p.G613R	1	0	1	8	3.18312E-05	D(0.001)	D(-5.83)	D(1.0)	30	0.507	
chr08	*CPSF1*	145623751	c.1835T>C	p.I612T	1	0	1	/	/	D(0.005)	D(-3.61)	D(0.971)	26.7	0.608	
chr08	*CPSF1*	145623949	c.1718G>A	p.G573E	1	0	1	/	/	D(0.024)	N(-1.83)	P(0.872)	23.2	0.28	
chr08	*CPSF1*	145623991	c.1676C>T	p.P559L	1	0	1	3	9.56328E-05	D(0.012)	N(-2.17)	B(0.007)	15.46	0.119	
chr08	*CPSF1*	145624009	c.1658A>T	p.E553V	1	0	1	/	/	D(0.047)	N(-2.38)	B(0.189)	23.4	0.103	
chr08	*CPSF1*	145624021	c.1646A>G	p.N549S	4	0	4	6	2.12483E-05	T(0.696)	N(-0.13)	B(0.0)	4.441	0.018	
chr08	*CPSF1*	145624180	c.1627C>T	p.R543C	1	0	1	5	0.000018171	D(0.046)	N(-2.04)	D(0.981)	25	0.159	
chr08	*CPSF1*	145624195	c.1612G>C	p.V538L	2	0	2	3	1.21036E-05	T(0.089)	N(-2.32)	P(0.938)	25.6	0.168	
chr08	*CPSF1*	145624552	c.1433C>T	p.A478V	12	0	12	16	6.26846E-05	T(1)	N(0.46)	B(0.085)	15.63	0.207	
chr08	*CPSF1*	145624697	c.1361C>T	p.S454L	1	0	1	2	8.02227E-06	T(0.192)	D(-2.92)	D(0.989)	25.5	0.2	
chr08	*CPSF1*	145624745	c.1313C>T	p.P438L	1	0	1	15	5.33375E-05	T(0.866)	N(-0.62)	B(0.0)	15.36	0.212	
chr08	*CPSF1*	145624866	c.1270G>A	p.V424M	5	0	5	7	2.49636E-05	T(0.081)	N(-0.84)	B(0.034)	21.2	0.179	

chr08	*CPSF1*	145624983	c.1237G>A	p.D413N	2	0	2	38	0.000139144	T(0.307)	N(-0.31)	P(0.924)	20.8	0.114	
chr08	*CPSF1*	145625060	c.1160G>A	p.R387H	6	1	5	8	3.32734E-05	D(0)	D(-4.69)	D(0.996)	32	0.352	
chr08	*CPSF1*	145625082	c.1138G>A	p.G380R	1	0	1	4	1.67531E-05	D(0.037)	D(-4)	D(0.989)	24.6	0.469	
chr08	*CPSF1*	145625214	c.1076C>T	p.A359V	1	0	1	5	2.61876E-05	T(0.15)	N(-2.14)	B(0.06)	14.77	0.069	
chr08	*CPSF1*	145625418	c.995A>G	p.Y332C	1	0	1	/	/	T(0.183)	D(-3.12)	P(0.90)	24.2	0.221	
chr08	*CPSF1*	145625440	c.973G>A	p.A325T	2	0	2	3	1.23001E-05	T(0.094)	N(-1.87)	P(0.638)	20.9	0.216	
chr08	*CPSF1*	145625560	c.937C>G	p.R313G	2	0	2	/	/	T(0.179)	D(-4.25)	B(0.116)	20.2	0.14	
chr08	*CPSF1*	145625605	c.892G>A	p.V298M	1	1	0	/	/	D(0.003)	N(-2.24)	D(0.999)	24.6	0.233	
chr08	*CPSF1*	145625650	c.847G>A	p.V283I	2	0	2	2	7.33181E-06	D(0.039)	N(-0.82)	D(0.979)	27.2	0.156	
chr08	*CPSF1*	145625656	c.841T>G	p.F281V	1	0	1	7	2.53975E-05	T(0.25)	D(-2.89)	B(0.213)	23.3	0.24	
chr08	*CPSF1*	145626088	c.663T>G	p.F221L	1	0	1	/	/	D(0.014)	D(-4.23)	P(0.943)	19.03	0.267	
chr08	*CPSF1*	145626123	c.628C>T	p.H210Y	1	0	1	/	/	T(0.069)	D(-3.91)	B(0.02)	19.99	0.195	
chr08	*CPSF1*	145626141	c.610A>G	p.I204V	14	2	12	6	2.40408E-05	T(0.173)	N(-0.56)	B(0.002)	5.309	0.042	
chr08	*CPSF1*	145626174	c.577G>T	p.V193L	1	0	1	1	4.00279E-06	T(1)	N(1.01)	B(0.002)	14.96	0.253	
chr08	*CPSF1*	145626174	c.577G>A	p.V193M	1	0	1	15	5.33424E-05	D(0.007)	N(-0.22)	P(0.63)	22.4	0.286	
chr08	*CPSF1*	145626438	c.419G>A	p.R140Q	13	6	7	235	0.000861437	T(0.255)	N(-0.13)	B(0.253)	16.43	0.08	
chr08	*CPSF1*	145626616	c.376C>G	p.P126A	1	0	1	7	3.75319E-05	T(0.288)	N(-1.49)	B(0.0)	19.81	0.074	
chr08	*CPSF1*	145626664	c.328C>T	p.P110S	1	0	1	5	3.07072E-05	D(0.013)	D(-4.46)	P(0.885)	23.6	0.31	
chr08	*CPSF1*	145626667	c.325G>A	p.D109N	1	0	1	1	6.08872E-06	T(0.075)	D(-3.04)	P(0.931)	23.7	#NNAA	
chr08	*CPSF1*	145626832	c.298G>A	p.D100N	1	0	1	/	/	D(0.005)	D(-2.99)	D(0.993)	25.1	0.168	
chr08	*CPSF1*	145626889	c.241A>G	p.M81V	1	0	1	11	3.90642E-05	D(0.01)	N(-2.38)	B(0.112)	20.9	0.239	
chr08	*CPSF1*	145626931	c.199G>A	p.E67K	1	0	1	4	1.42605E-05	D(0.009)	D(-3.08)	D(0.964)	23.8	0.277	
chr08	*CPSF1*	145634494	c.49T>A	p.S17T	1	0	1	/	/	T(0.139)	N(-1.32)	B(0.053)	9.883	0.219	
chr08	*CPSF1*	145634528	c.15C>G	p.Y5*	1	1	0	/	/	NA	NA	NA	36	NA	Ouyang JM, et al.

chr06	*TNFRSF21*	47200589	c.1880G>A	p.R627Q	1	0	1	3	1.19325E-05	T(0.095)	N(-1.03)	B(0.02)	23.5	0.123
chr06	*TNFRSF21*	47200631	c.1838G>A	p.R613Q	4	0	4	3	1.0609E-05	D(0.005)	N(-0.39)	B(0.079)	23.4	0.149
chr06	*TNFRSF21*	47200703	c.1766T>G	p.V589G	20	3	17	/	/	D(0.000)	N(-2.27)	D(0.999)	27.4	0.628
chr06	*TNFRSF21*	47202424	c.1720G>A	p.G574S	4	0	4	5	1.82498E-05	D(0.029)	N(-1.94)	D(0.999)	29.7	0.502
chr06	*TNFRSF21*	47202496	c.1648T>C	p.F550L	1	0	1	4	1.44877E-05	D(0)	N(-1.95)	P(0.837)	25.5	0.561
chr06	*TNFRSF21*	47202537	c.1607T>A	p.L536Q	1	0	1	1	4.69096E-06	D(0.001)	N(-1.42)	B(0.23)	22.6	0.332
chr06	*TNFRSF21*	47202564	c.1580A>G	p.N527S	1	0	1	180	0.000892769	T(0.304)	N(-0.38)	B(0.001)	4.572	0.115
chr06	*TNFRSF21*	47221033	c.1468G>A	p.V490I	1	0	1	1	3.99974E-06	D(0)	N(-0.36)	P(0.685)	26.2	0.435
chr06	*TNFRSF21*	47221096	c.1405C>T	p.R469W	1	0	1	7	2.78976E-05	D(0)	D(-2.85)	D(0.987)	32	0.815
chr06	*TNFRSF21*	47221146	c.1355A>G	p.Y452C	1	0	1	2	7.9552E-06	D(0.044)	D(-3.54)	B(0.152)	23.6	0.657
chr06	*TNFRSF21*	47221225	c.1276G>C	p.V426L	1	0	1	2	7.98E-06	D(0.002)	N(-0.99)	P(0.498)	25.1	0.476
chr06	*TNFRSF21*	47251754	c.1163T>C	p.I388T	1	0	1	1	3.97719E-06	D(0.044)	N(-2.09)	B(0.043)	24.1	0.457
chr06	*TNFRSF21*	47251873	c.1044G>C	p.E348D	1	0	1	28	9.90211E-05	D(0)	N(-0.77)	D(0.994)	24.1	0.369
chr06	*TNFRSF21*	47252055	c.862G>A	p.V288M	4	0	4	235	0.0008758	T(0.525)	N(0.27)	B(0.002)	0.49	0.062
chr06	*TNFRSF21*	47252061	c.856G>A	p.E286K	1	1	0	2	8.54139E-06	D(0.045)	N(-0.49)	B(0.005)	19.61	0.11
chr06	*TNFRSF21*	47252157	c.760A>G	p.T254A	2	0	2	1	5.62924E-06	T(0.071)	N(-1.11)	B(0.15)	18.37	0.129
chr06	*TNFRSF21*	47253739	c.689T>C	p.F230S	1	0	1	/	/	T(0.179)	N(-0.42)	B(0.001)	15.23	0.029
chr06	*TNFRSF21*	47253757	c.671C>G	p.S224C	2	0	2	/	/	D(0.001)	N(-0.77)	B(0.319)	18.46	0.101
chr06	*TNFRSF21*	47253779	c.649T>G	p.F217V	1	0	1	/	/	T(0.174)	N(-1.09)	B(0)	5.701	0.022
chr06	*TNFRSF21*	47253823	c.605C>T	p.P202L	1	0	1	4	1.59095E-05	T(0.092)	D(-4.68)	B(0.002)	21.6	0.102
chr06	*TNFRSF21*	47253832	c.596T>G	p.V199G	1	0	1	5	1.99283E-05	D(0)	D(-5.14)	B(0.282)	25.8	0.354
chr06	*TNFRSF21*	47254004	c.424G>A	p.A142T	3	0	3	5	1.99445E-05	T(0.228)	N(-0.23)	B(0)	16.27	0.145
chr06	*TNFRSF21*	47254136	c.292A>T	p.R98W	3	1	2	5	1.99445E-05	D(0)	D(-4.01)	D(0.915)	29.2	0.747
chr06	*TNFRSF21*	47254169	c.259G>A	p.V87I	1	1	0	17	6.02486E-05	T(0.075)	N(-0.83)	B(0.088)	22.7	0.227

chr06	*TNFRSF21*	47254253	c.175C>T	p.R59C	1	1	0	12	4.24845E-05	D(0.008)	N(-2.21)	P(0.685)	27	0.608
chr06	*TNFRSF21*	47254302	c.126G>C	p.Q42H	1	0	1	/	/	D(0.012)	N(-1.86)	P(0.887)	23.2	0.212
chr06	*TNFRSF21*	47254323	c.105C>A	p.F35L	1	0	1	/	/	T(0.169)	N(-0.55)	B(0.001)	16.86	0.037
chr06	*TNFRSF21*	47277195	c.53C>G	p.A18G	2	0	2	/	/	D(0.01)	N(-0.42)	B(0.011)	13.17	0.028

Abbreviations: / indicated none in the gnomAD database; B = benign; D = damage; N = neutral; NA = not applicable; P = probably damage; T = tolerant.