

Variants of the MAPT H2/H2 diplotype

Locus	Genotype	Ref	Observed	Type	Genes	% Frequency	Exon
chr17:44039691	G/G	A	G	SNV	MAPT	100.00	2
chr17:44060775	T/T	C	T	SNV	MAPT	99.55	6
chr17:44061023	A/A	G	A	SNV	MAPT	99.87	6
chr17:44061036	C/C	T	C	SNV	MAPT	99.82	6
chr17:44061278	T/T	C	T	SNV	MAPT	95.28	6
chr17:44067400	C/C	T	C	SNV	MAPT	93.39	8
chr17:44068924	A/A	G	A	SNV	MAPT	99.93	9
chr17:44071294	C/C	T	C	SNV	MAPT	100.00	10
chr17:44073889	G/G	A	G	SNV	MAPT	99.92	11
chr17:44073973	C/C	T	C	SNV	MAPT	100.00	11
chr17:44101563	C/C	T	C	SNV	MAPT	99.87	15
chr17:44101871	G/G	A	G	SNV	MAPT	100.00	15
chr17:44102443	CCT/CCT	C	CCT	INDEL	MAPT	98.56	15
chr17:44102604	C/C	T	C	SNV	MAPT	99.83	15
chr17:44102638	G/G	A	G	SNV	MAPT	99.97	15
chr17:44102682	CT/CT	C	CT,T	INDEL	MAPT	99.35, T=0	15
chr17:44102689	C/C	G	C	SNV	MAPT	100.00	15
chr17:44102741	T/T	TCA	T	INDEL	MAPT	100.00	15
chr17:44102865	C/C	A	C	SNV	MAPT	99.52	15
chr17:44102933	C/C	T	C	SNV	MAPT	100.00	15
chr17:44103296	C/C	T	C	SNV	MAPT	97.51	15
chr17:44103616	T/T	C	T	SNV	MAPT	99.85	15
chr17:44103825	CA/CA	TG	CA	MNV	MAPT	99.97	15
chr17:44104278	T/T	TG	T	INDEL	MAPT	100.00	15
chr17:44104343	C/C	A	C	SNV	MAPT	99.85	15
chr17:44104411	C/C	CTCC	C	INDEL	MAPT	100.00	15
chr17:44104509	C/C	T	C	SNV	MAPT	99.97	15
chr17:44104576	C/C	CCTT	C	INDEL	MAPT	99.95	15
chr17:44105395	G/G	A	G	SNV	MAPT	99.90	15

Coding	Amino Acid Change	Variant Effect	dbSNP	ClinVar	Coverage	MAF
c.-13A>G	p.?	unknown	rs17650901	Benign	254	0.086
c.605C>T	p.Pro202Leu	missense	rs63750417	Benign	3996	0.088
c.853G>A	p.Asp285Asn	missense	rs62063786	Benign	3996	0.088
c.866T>C	p.Val289Ala	missense	rs62063787	Benign	3965	0.088
c.1108C>T	p.Arg370Trp	missense	rs17651549	Benign	1018	0.086
c.1339T>C	p.Ser447Pro	missense	rs10445337	Benign	2223	0.088
c.1479G>A	p.(=)	synonymous	rs1052551	Benign	1340	0.086
c.1512T>C	p.(=)	synonymous	rs62063845	Benign	3993	0.088
c.1686A>G	p.(=)	synonymous	rs1052553	Benign	3993	0.088
c.1770T>C	p.(=)	synonymous	rs17652121	Benign	3999	0.088
c.2357T>C	p.?	unknown	rs9468	Benign	3971	0.086
c.2665A>G	p.?	unknown	rs8712	Benign	1259	0.086
c.3237_3238insCT	p.?	unknown	rs113598111,rs113598112	Benign	139	0.086
c.3398T>C	p.?	unknown	rs1052587	Benign	2888	0.086
c.3432A>G	p.?	unknown	rs1052590	Benign	3983	0.086
c.3476_3477insT	p.?	unknown	rs5820605	Benign	3989	0.382
c.3483G>C	p.?	unknown	rs1052594	Benign	3994	0.086
c.3536_3537delCA	p.?	unknown		Benign	3978	
c.3659A>C	p.?	unknown	rs17574040	Benign	3993	0.086
c.3727T>C	p.?	unknown	rs16940799	Benign	3995	0.086
c.4090T>C	p.?	unknown	rs7687	Benign	3818	0.086
c.4410C>T	p.?	unknown	rs17652748	Benign	3992	0.086
c.4619_4620delTGinsCA	p.?	unknown	rs16940806,rs7711705	VUS	3984	0.086
c.5073delG	p.?	unknown	rs5820606	Benign	3671	0.086
c.5137A>C	p.?	unknown	rs2158257	Benign	3320	0.086
c.5206_5208delTCC	p.?	unknown	rs199706121,rs568475466		2364	0.086
c.5303T>C	p.?	unknown	rs17574228	Benign	3985	0.086
c.5371_5373delCTT	p.?	unknown	rs760422692	Benign	3972	0.174
c.6189A>G	p.?	unknown	rs7521	Benign	3999	0.386 (1)

0.386 (ref)