TPMT allele nomenclature

The table defines all the single-nucleotide polymorphisms (SNPs) in *TPMT* as of May 2019

Allele	dbSNP rsID and corresponding nucleotides on the positive chromosomal strand (for standardization) and ClinVar accession number if applicable	Nucleotide changes in the *TPMT* gene (given on the negative chromosomal strand, NCBI reference sequence NM_000367.2)	Gene location	Amino acid Change (NCBI reference sequence NP_000358.1)	References
*TPMT*1*	rs2842934 allele A[1]	Wild type 474T[1]			
*TPMT*1A*	ND, G>A	-178C>T	Exon I		[1]
*TPMT*1S*	A>G at rs2842934	474T>C	Exon VII	Ile158Ile	[2]
*TPMT*2*	C>G at rs1800462	238G>C	Exon V	Ala80Pro	[3, 4, 5, 6, 7]
*TPMT*3A*	C>T at rs1800460 T>C at rs1142345	460G>A 719A>G	Exon VII Exon X	Ala154Thr Tyr240Cys	[4, 5, 6, 7, 8]
*TPMT*3B*	C>T at rs1800460	460G>A	Exon VII	Ala154Thr	[4, 6, 7, 8]
*TPMT*3C*	T>C at rs1142345	719A>G	Exon X	Tyr240Cys	[4, 6, 7, 8]
*TPMT*3D*	C>A at s72552739 C>T at rs1800460 T>C at rs1142345	292G>T 460G>A 719A>G	Exon V Exon VII Exon X	Glu98Stop Ala154Thr Tyr240Cys	[9]
*TPMT*3E*	A>T at rs3931660 T>A at rs12529220 A>G at rs2518463 C>T at rs1800460 A>G at rs2842934 T>C at rs1142345	140+114T>A 141-101A>T 366+58T>C 460G>A 474T>C 719A>G	Intron III Intron III Intron IV Exon VII Exon VII Exon X	 Ala154Thr Ile158Ile Tyr240Cys	[10]
*TPMT*4*	C >T at rs1800584	626-1G>A	Intron IX/ exon X in splice junction		[9, 11]

*TPMT*5*	A>G at rs72552740	146T>C	Exon IV	Leu49Ser	[4, 6, 9]
*TPMT*6*	T>A at rs75543815	539A>T	Exon VIII	Tyr180Phe	[4, 6, 9]
*TPMT*7*	A>C at rs72552736	681T>G	Exon X	His227Gln	[1, 4, 6, 9]
*TPMT*8*	C>T at rs56161402	644G>A	Exon X	Arg215His	[4, 6, 12]
*TPMT*9*	T>G at rs151149760	356A>C	Exon V	Lys119Thr	[4, 6, 13]
*TPMT*10*	C>G at rs72552737	430G>C	Exon VII	Gly144Arg	[4, 6, 14, 15]
*TPMT*11*	C>T at rs72552738	395G>A	Exon VI	Cys132Tyr	[4, 6, 16]
*TPMT*12*	G>A at rs200220210	374C>T	Exon VI	Ser125Leu	[4, 6, 15]
*TPMT*13*	T>A at rs72552742	83A>T	Exon III	Glu28Val	[4, 6, 15]
*TPMT*14*	T>C at rs9333569	1A>G	Exon III	Met1Val	[6, 17]
*TPMT*15*	C>T at rs9333570	495-1G>A	Intron VII/ exon VIII in splice junction		[17]
*TPMT*16*	C>T at rs144041067	488G>A	Exon VII	Arg163His	[6, 13, 18]
*TPMT*17*	ND, G>C	124C>G	Exon III	Gln42Glu	[6, 13]
*TPMT*18*	C>T at rs777686348	211G>A	Exon IV	Gly71Arg	[6, 13]
*TPMT*19*	ND, T>G	365A>C	Exon V	Lys122Thr	[6, 18]
*TPMT*20*	T>C at rs150900439	712A>G	Exon X	Lys238Glu	[6, 19]
*TPMT*21*	G>C at rs200591577	205C>G	Exon IV	Leu69Val	[6, 19]
*TPMT*22*	ND, C>G	488G>C	Exon VII	Arg163Pro	[6, 19]
*TPMT*23*	G>C at rs74423290	500C>G	Exon VIII	Ala167Gly	[20]
*TPMT*24*	C>A at rs6921269	537G>T	Exon VIII	Gln179His	[21]
*TPMT*25*	A>G at rs377085266	634T>C	Exon X	Cys212Arg	[21]
*TPMT*26*	A>G at rs72556347	622T>C	Exon IX	Phe208Leu	[22]
*TPMT*27*	ND, A>C	319T>G	Exon V	Tyr107Asp	[23]
*TPMT*28*	ND, C>G	349G>C[2]	Exon V	Gly117Arg	[24]
*TPMT*29*	A>G at rs267607275	2T>C	Exon III	Met1Thr	[25]
*TPMT*30*	Old TPMT*20/*24, C>T at rs750424422	106G>A	Exon III	Gly36Ser	[6, 26]
*TPMT*31*	Old TPMT*28 A>G at rs79901429	611T>C	Exon IX	Ile204Thr	[27]
*TPMT*32*	C>T at rs115106679	340G>A	Exon V	Glu114Lys	[28]
*TPMT*33*	G>A at rs112339338	487C>T	Exon VII	Arg163Cys	[28]
*TPMT*34*	G>A at rs111901354	244C>T	Exon V	Arg82Trp	[28]

*TPMT*35*	ND	200T>C[3]	Exon III	Phe67Ser	[29]
*TPMT*36*	ND	595G>A[3]	Exon VIII	Val199Ile	[29]
*TPMT*37*	A>T at rs398122996	648T>A	Exon X	Cys216Ter	[30]
*TPMT*38*	G>A at rs772832951	514T>C	Exon VIII	Ser172Pro	[31]
*TPMT*39*	A>G at rs281874771	218C>T	Exon VI	Ala73Val	[32]
*TPMT*40*	T>C at rs139392616	677G>A	Exon X	Arg226Q	[33]
*TPMT*41*	T>G at rs1142345	719A>C	Exon X	Tyr240Ser	[34]
*TPMT*42*	ins A at rs759836180, SCV000778513	95_96insA	Exon III	Lys32LysfsX58	[35]
*TPMT*43*	SCV000778514 rs753545734, SCV000778515	234G>T 262G>A	Exon V Exon V	Trp78Cys Gly88Ser	[35]
*TPMT*44*	rs201695576, SCV000778516	497A>G	Exon VIII	Tyr166Cys	[35]

ND – Not reported to dbSNP, [1]dbSNP reports G>A at this position: however the TPMT nomenclature committee has defined wildtype as having allele A at this position (positive chromosomal strand) and the *1S allele as having allele G at this position (positive chromosomal strand): [2] incorrect nucleotide substitutions given in reference 24: the corrected nucleotide substitution is included in the table (personal communication T. Marinaki).[3] These alleles was numbered differently in the original publication [29].

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