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TPMT*V4 Allele

National Cancer Institute

Source

National Cancer Institute. *TPMT*V4 Allele*. NCI Thesaurus. Code C45797.

TPMT*V4 Allele is a variant form of 34-kb 10-exon human TPMT Gene (TPMT Family), which encodes polymorphic monomeric 245-aa 28-kDa cytoplasmic Thiopurine S-Methyltransferase. Inhibited by S-adenosyl-L-homocysteine, TPMT catalyzes thiopurine S-methylation, an important metabolic pathway for thiopurine drugs such as 6-mercaptopurine. TPMT contains a variable number of 17/18 bp tandem repeats (VNTR), from 3-9 (*V3-*V9), within the GC-rich 5'-flanking TPMT promoter region that modulate the level of TPMT enzyme activity (to a smaller extent than ORF-based SNP effects). A decrease in gene expression is seen with increasing repeat numbers. TPMT*V4 contains 4 VNTR. TPMT activity is thought to be an important determinant of toxicity associated with thiopurine medications.