

THYROPURINE METHYL TRANSFERASE (TPMT) GENOTYPING

THYROPURINE METHYL TRANSFERASE (TPMT) GENOTYPING (Real Time PCR)	
TPMT*1 wild type	Detected/ Not Detected
TPMT*2 G238C	Detected/ Not Detected
TPMT*3A G460A and A719G	Detected/ Not Detected
TPMT*3B G460A	Detected/ Not Detected
TPMT*3C A719G	Detected/ Not Detected

Interpretation

GENOTYPE	EFFECT OF POLYMORPHISM
TPMT* 1 Wild Type	Normal enzyme activity
TPMT* 2 G238C	Reduced enzyme activity
TPMT* 3A G460 & A719G	No enzyme activity
TPMT* 3B G460A	Reduced enzyme activity
TPMT* 3C A719G	Reduced enzyme activity

Note:

1. This test predicts the TPMT enzyme activity based on polymorphism at three positions 238, 460 and 719 of TPMT gene
2. Current TPMT phenotype may not be reproducible in patients who have received blood transfusions within 30-60 days of testing
3. This is an in-house developed test for Research purposes only. Results to be interpreted in context of clinical findings, and other laboratory data.

Comments

Measurement of TPMT activity is advised prior to commencing the treatment of patients with Thiopurine drugs such as Azathioprine, 6-Mercaptopurine and 6-Thioguanine. Patients with low activity (10% prevalence) or especially absent activity (0.3% prevalence) are at a heightened risk of drug-induced bone marrow toxicity due to accumulation of the unmetabolised drug. About 5% of all Thiopurine therapies will fail due to toxicity. There appears to be a great deal of

variation in TPMT mutation, with ethnic differences in mutation types accounting for variable responses to 6MP.

Dr Lal PathLabs