

Thiopurine S methyltransferase

General:

Thiopurine S-methyltransferase deficiency (TPMT-deficiency) is an autosomal recessive trait associated with severe hematopoietic toxicity when patients are treated with standard doses of the antineoplastic agents. Thiopurine methyltransferase (TPMT) catalyzes the S-methylation of thiopurine drugs such as 6-mercaptopurine (6-MP), thioguanine and azathioprine (AZA). These drugs are used to treat conditions such as acute lymphoblastic leukemia, inflammatory bowel disease, rheumatoid arthritis, and organ transplant rejection. The TPMT gene exhibits significant genetic polymorphisms among all ethnic groups studied.

Patients who inherited very low levels of TPMT activity are at greatly increased risk for thiopurine-induced toxicity such as myelosuppression, when treated with standard doses of these drugs, while subjects with very high activity may be undertreated. Moreover, clinical drug interactions may occur due to TPMT induction or inhibition. Identification of the TPMT mutant alleles allows physicians to adjust the dosage of the thiopurine drugs to the genotype of the patient or to use alternatives, improving therapeutic outcome.

The following tests are available:

- **TPMT enzyme activity in RBC**

Material: 3 ml EDTA blood

TAT: 7-10 days*

Method: HPLC

Units: nmol/g Hb*h

Ref.- range: normal >20

Heterozygous 10-20

Homozygous <10

- **TPMT genetic test**

Material: 2 ml EDTA blood

TAT: 7-10 days*

Method: PCR

Ref.- range: see report

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit

<http://www.fml-dubai.com/parameter-listings/>