MAYO CLINIC
Mayo Medical Laboratories

REFERENCE VALUE CHANGE REFERRAL
NOTIFICATION DATE: January 23, 2012
EFFECTIVE DATE: Immediately

PROMETHEUS TPMT GENETICS
Test ID: FPRTG
Secondary ID: 91565

EXPLANATION: Effective immediately, Test ID FPRTG, referred to Prometheus Laboratories, Inc., will reflect the following change:

CURRENT REFERENCE VALUE:
Reference Range: TPMT*1/TPMT*1

Prometheus TPMT Genetics is an analysis to determine an ability to produce thiopurine methyltransferase (TPMT) activity. It is a method to identify patients at risk for acute toxicity from 6-MP or azathioprine. This profile provides a breakdown of a patient's genetics. The distribution of TPMT activity is trimodal; homozygous normal (89%), heterozygous (11%), and homozygous recessive (0.3%). Approximately 1 in 1213 individuals may have a low TPMT enzyme activity (homozygous low) resulting from known and theoretical mutations that are not included in this panel.

Notes: Genetic testing results are reported above as the individual allele present on each chromosome for three different polymorphisms; G238C, G460A, and A719G within the TPMT gene on chromosome 6. The alleles are numbered based on order of discovery.

A combination of Cepheid Smart Mix Reagents with ABI (Applied Biosystems Sequence Detection System) Prism 7000 allelic discrimination was used in determining the presence or absence of 3 polymorphisms of the TPMT gene located on chromosome 6. Included are 3 separate PCR reactions, 3 different sets of probes and primers. This test was developed and its performance characteristics determined by Prometheus Laboratories Inc. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA).The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research.This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing.

The homozygous recessive genotype predicts a deficient capacity to produce TPMT enzyme activity. TPMT enzyme activity is essential for normal metabolism of azathioprine or 6-mercatopurine.

Our genotyping procedures will not distinguish between TPMT*1/TPMT*3A from the rare TPMT*3B/TPMT*3C which has a frequency of 1:120,890. This rare genotype is associated with low enzyme activity. Enzyme activity evaluation or sequencing is necessary to definitively identify this rare genotype.

NEW REFERENCE VALUE:
Reference Range: TPMT*1/TPMT*1

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NOTE: This change may impact test set-up information and could require a change to file definition. Please review the Test Set-Up information for specifics at http://www.mayomedicallaboratories.com/test-notifications/index.html.

QUESTIONS: Contact Mary Erath, MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710