MAYO CLINIC Mayo Medical Laboratories

TEST DEFINITION

1/23/2012

CODE

CODE

ORDER RESULT PERFORM

CHECKING NORMALS

SITE *

PRINT NORMALS

FPRTG PROMETHEUS TPMT GENETICS Y093

TRANSPORT TEMP : REFRIGERATED <30 DAYS\AMBIENT <8 DAYS

Z1518PROMETHEUS TPMT GENETICS

TITLE

UNITS:

TEST CODE ALWAYS MESSAGE - [TZ355] T7355 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 7500Fast allelic discrimination was used in determining THE PRESENCE OR ABSENCE OF 3 POLYMORPHISMS OF THE TPMT GENE

> > TEST SETUP INFORMATION

ORDER RESULT PERFORM CODE CODE TITLE SITE * CHECKING NORMALS PRINT NORMALS

FPRTG (CONTINUED...)

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.

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-mercaptopurine.

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.

*PERFORMING SITE LEGEND

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