

**PRO-Predict<sup>®</sup> TPMT Genetic Assessment**
**Patient & Order Information**

 Order ID **114846**  
 Patient **Test, Patient**  
 DOB **01/10/1900**  
 SSN **123-45-6789** Sex **F**  
 Institution ID Promethus ID **21740**  
 Ordered 01/05/2001 Completed 05/19/2003  
 Ordered By Example Physician M.D.  
 ICD9 Codes 789.00

**Report Recipient**

 Example Physician M.D.  
 Prometheus Laboratories Inc.  
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 Suite 123  
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 USA  
 858/824-0895 Phone 858/824-0896 Fax

Sample ID: SC01050042 Collection Date: 01/04/2001 (Whole Blood) Institution Sample ID:

**Test Results**

Assay Name	Result	Reference Range
<b>PRO-Predict<sup>®</sup> TPMT Genetic Assessment</b>	<b>TPMT*1/TPMT*1</b>	<b>TPMT*1/TPMT*1</b>

**Alleles present are associated with NORMAL ENZYME ACTIVITY.**

Report Reviewed by the Laboratory Medical Director.

**PRO-Predict<sup>®</sup> TPMT Genetic Assessment** 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%) (1). 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 within the TPMT gene on chromosome 6. The alleles are numbered based on order of discovery.

TaqMan<sup>™</sup> (Applied Biosystems Sequence Detection System) allelic discrimination PCR methodology 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.

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 (2).

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.

**References:**

- (1) Lennard, L. et al., "The Clinical Pharmacology of 6-Mercaptopurine", European Journal of Clinical Pharmacology, Vol.43, 1992, p 329-339.
- (2) Charles R. Yates et al., "Molecular Diagnosis of Thiopurine S-Methyltransferase Deficiency: Genetic Basis for Azathioprine and Mercaptopurine Intolerance", Annals of Internal Medicine, Vol. 126, No. 8, April 1997, p 608-614.