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Your Test Results

Case Number:	**18020500**
Patient Name:	**Aprana**
Age/Sex:	**17 Yrs/Female**
Patient Location:	**Ambala**
Hospital Name:	**KOS Lab**
Physician Name:	**Dr. Yugam Chopra**
Date & Time of Accessioning:	**02/05/2018 10:27 Hrs**
Date & Time of Reporting:	**07/05/2018 16:56 Hrs**

TEST NAME

TPMT genotyping study

SPECIMEN INFORMATION

EDTA peripheral blood collected on 30/04/2018 at 18:00 Hrs

CLINICAL HISTORY

Not Provided

METHODOLOGY

PCR-Sequencing

DIAGNOSIS

MOLECULAR TEST	RESULT
TPMT Genotyping Study By PCR-Sequencing	**TPMT*1/*1 Genotype Detected**

INTERPRETATION OF RESULTS

Individuals with 2 variant alleles have low or no TPMT activity, while those with 1 variant allele have intermediate TPMT activity. Wild-type (TPMT*1) homozygote, on the other hand, have normal enzyme activity.

TPMT Alleles Tested	Genetic Variant	dbSNP	Predicted Enzyme Activity
TPMT *1	None	---------	Normal
TPMT *2	c.238G>C	rs1800462	Non Functional
TPMT *3A	c.460G>A and c.719A>G	rs1800460 and rs1142345	Non Functional
TPMT *3B	c.460G>A	rs1800460	Non Functional
TPMT *3C	c.719A>G	rs1142345	Non Functional

COMMENT

Clinical Background: Thiopurine drugs (azathioprine, 6-mercaptopurine, and 6-thioguanine) are used to treat patients with leukemia, rheumatic disease, inflammatory bowel disease, or solid organ transplant. These drugs must be metabolized to 6-thioguanine nucleotides (6-TGN) for activity. Thiopurine methyltransferase (TPMT) plays an active role in this drug metabolism pathway and suboptimal activity of TPMT results in impaired drug metabolism causing adverse drug reactions or lack of therapeutic response. Polymorphisms in the *TPMT* gene results in reduced enzymatic activity. *TPMT*2* (238G>C), *TPMT*3A* (460G>A and 719A>G), *TPMT*3B* (460G>A), and *TPMT*3C* (719A>G) accounts for reduced TPMT activity in >95% cases. Patients without a wild-type allele of *TPMT* gene are at risk of severe haematological toxicities is standard dosages of thiopurine medications are administered to them.

Dr. Rahul Katara, Ph.D., Molecular Scientist

CŌRE DIAGNOSTICS™

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LIMITATION

This assay screens only the above mentioned alleles in *TPMT* gene. In cases where none of the variant alleles are detected, a wild type allele (*1) is reported. Kindly note this does not rule out absence of rear *TPMT* genetic variants not screened in this test. This test covers more than 95 percent of non- functional alleles for the tested population. Drug metabolism may be affected by non-genetic factors. Rare diagnostic errors may occur due to primer-site mutations.

REFERENCES

1. McLeod HL, Siva C. The thiopurine S-methyltransferase gene locus–implications for clinical pharmacogenomics. Pharmacogenomics. 2002; 3:89-98.
2. Evans WE. Pharmacogenetics of thiopurine S-methyltransferase and thiopurine therapy. The Drug Monit. 2004; 26:186-191.
3. Yates CR, Krynetski EY, Loennechen T, et al. Molecular diagnosis of thiopurine S-methyltransferase deficiency: genetic basis for azathioprine and mercaptopurine intolerance. Ann Intern Med. 1997; 126:608-614.
4. Black AJ, McLeod HL, Capell HA, et al. Thiopurine methyltransferase genotype predicts therapy-limiting severe toxicity from azathioprine. Ann Intern Med. 1998; 129:716-718.
5. Kurzawski M, Dziewanowski K, Gawronska-Szklarz B, et al. The impact of thiopurine S-methyltransferase polymorphism on azathioprine-induced myelotoxicity in renal transplant recipients. The Drug Monitor. 2005; 27:435-441.

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Questions?

Contact us at **+91 124 4615 615**

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CONDITIONS OF REPORTING

1. The tests are carried out in the lab with the presumption that the specimen belongs to the patient named or identified in the bill/test request form.
2. The test results relate specifically to the sample received in the lab and are presumed to have been generated and transported per specific instructions given by the physicians/laboratory.
3. The reported results are for information and are subject to confirmation and interpretation by the referring doctor.
4. Some tests are referred to other laboratories to provide a wider test menu to the customer.
5. CORE Diagnostics Pvt. Ltd. shall in no event be liable for accidental damage, loss, or destruction of specimen, which is not attributable to any direct and mala fide act or omission of CORE Diagnostics Pvt. Ltd. or its employees. Liability of CORE Diagnostics Pvt. Ltd. for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.

This is a transcribed report and the test was performed at the laboratory OSL 14.