DUPLICATE 07/05/2025 10:56:19

# TAN TOCK SENG HOSPITAL

11 Jalan Tan Tock Seng, Singapore 308433

## MOLECULAR DIAGNOSTIC LABORATORY

## FINAL REPORT

| | | | |
|---|---|---|---|
| Lab Accession No. | : 5053006148 | Location | : TEST LOCATION |
| Name | : TEST PAT | Race | : Chinese |
| NRIC | : 00000000TEST | Sex | : M |
| DOB | : 01/01/2002 | Age | : 23Y |
| Requested By | : TESTING DOCTOR | Date Received | : 07/05/2025 10H38M |
| Comments | : | Date Report | : 07/05/2025 10H41M |

### MOLECULAR (Test done in TTSH MDL)

**PGX Targeted Panel**

**Result**

| Gene | Genotype | Predicted Phenotype |
|---|---|---|
| **ABCG2 c.421:** | A/A | Poor function |
| **CYP2C19:** | *1/*3 | Intermediate metabolizer |
| **CYP2C9:** | *1/*1 | Normal metabolizer (AS:2) |
| **CYP2D6:** | *1/*1 | Normal metabolizer (AS:2) |
| **CYP3A5:** | *3/*3 | Poor metabolizer |
| **CYP4F2:** | *1/*3 | Intermediate metabolizer |
| **DPYD:** | *1/HapB3 | Intermediate metabolizer (AS:1.5) |
| **HLA-A*31:01:** | Negative | No increased risk of carbamazepine hypersensitivity |
| **HLA-B*15:02:** | Negative | No increased risk of carbamazepine hypersensitivity |
| **HLA-B*57:01:** | Positive | Increased risk of abacavir hypersensitivity |
| **HLA-B*58:01:** | Positive | Increased risk of allopurinol hypersensitivity |
| **NUDT15:** | *1/*1 | Normal metabolizer |
| **SLCO1B1:** | *1/*37 | Normal function |
| **TPMT:** | *1/*1 | Normal metabolizer |
| **UGT1A1:** | *28/*28 | Poor metabolizer |
| **VKORC1 -1639:** | G/G | Low warfarin sensitivity |

**Material Submitted**

Blood (EDTA)

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### Assay Information

The test is intended to provide information to help in making medication decisions. Using array-based technology (Thermo Fisher Scientific), the lab-developed assay tests for specific variants in ABCG2 (rs2231142), CYP2C9 (*2,*3), CYP2C19 (*2,*3,*17), CYP2D6 (*2,*4,*5,*8,*10,*14,*29,*34,*36,*39,*41,*42,*49,*53,*69,*70,*88,*107,*109,*114), CYP3A5 (*3), CYP4F2 (*3), DPYD (HapB3,*2A), HLA-A*31:01, HLA-B*15:02, HLA-B*57:01, HLA-B*58:01, NUDT15 (*2,*3,*6), SLCO1B1 (*5,*15,*37), TPMT (*3A,*3B,*3C), UGT1A1 (*6,*28) and VKORC1 (rs9923231). CYP2D6 copy number status was assessed at sites within 5'-flanking region and exon 9. The genotype, with associated star alleles, is assigned using standard allelic nomenclature as published by the Pharmacogene Variation (PharmVar) Consortium (<http://www.pharmvar.org/>). Phenotype is predicted based on the specific variants interrogated. Assignment of a *1 allele indicates that none of the targeted variants were detected, but does not rule out the presence of other variants in the gene. The activity score (AS) system is used for phenotype prediction of CYP2D6, CYP2C9 and DPYD.

This test was developed and its performance characteristics determined by the Molecular Diagnostic Laboratory (MDL), TTSH. It is used for clinical purposes and should not be regarded as investigational. MDL is accredited by College of American Pathologists (CAP) to perform high-complexity testing.

### Disclaimer

This test does not detect all variants of the gene tested. Non-detected variants, other genetic and/or non-genetic factors including, but not limited to, medical conditions and drug-drug interactions, may impact the phenotype. Absence of a detectable gene variant does not rule out the possibility that a patient has an ultrarapid, rapid, intermediate or poor metabolizer phenotype. Rare polymorphisms could lead to false positive or negative results. There is a possibility that the results will be uninterpretable. Assay performance below the minimum acceptable criteria or failed region will be noted.

The method used to genotype does not determine haplotype phasing. The most likely diplotype based upon observed frequencies will be reported. Furthermore, this assay is unable to distinguish HLA-B*15:02 from HLA-B*15:13. Decisions regarding prescription and treatment should not be solely based on this test and the information contained in this report. Drug-label guidance and clinical correlation are recommended.

Information contained within this report is dependent on the date of generation and/or database version used to generate that report. The requesting party of this test has requested for the interpretation of this test result to be excluded in the report, which is not standard practice. However, if the interpretation is subsequently required, an addendum can be provided for an administrative fee.

### Electronically signed by

Dr Ong Kiat Hoe, Senior Consultant Haematologist
Dr Goh Liuh Ling, Senior Principal Scientific Officer

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