Kemenkes
RSPON Mahar Mardjono

Address MT Haryono St No.Kav.11, Cawang, Kramat Jati, East Jakarta City, Jakarta 13630

Tel (021) 29373377

Laboratory No.	:	Diagnosis	:
Patient Name	:	Referring Clinician	:
DOB	:	Specimen Type	:
Sex	:	Specimen Collected	:
MR No.	:	Specimen Received	:
Unit	:	Test Finished	:
Test Requested	:		

PHARMACOGENOMICS REPORT

Patient Clinical Information

Examination Detail

Drug Name	Gene	Diplotype	Phenotype
Clopidogrel	*CYP2C19*		

Variants tested: *CYP2C19*: *2 (c.681G>A, rs4244285), *3 (c.636G>A, rs4986893), *17 (c.-806C>T, rs12248560)

Test Implication

Normal or increased clopidogrel active metabolite formation[1]

Recommendation

Based on Clinical Pharmacogenetics Implementation Consortium 2022 guideline (CPIC)[1]	If the patient is using clopidogrel, use clopidogrel in a standard dosage (75 mg/day)[1]

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NOTE

The recommendation of genotype—phenotype association (metabolizer profile) might be updated based on the clinical guideline. This result is relevant for patients aged ≥18 years old.

TEST PERFORMANCE SUMMARY

The genomic DNA was extracted from whole blood specimen using the Roche High Pure PCR Template Preparation Kit, following the manufacturer's protocol. Extracted gDNA were then subjected to amplification using a set of in-house primers targeting the *CYP2C19* gene's exonic regions. A tiling approach for the targeted regions were then resulting in amplicons of approximately 1.5 kbp in size. The amplicons were then barcoded, cleaned, and ligated with adapters in accordance with the library preparation protocol using the Rapid Barcoding Kit 96 V14 (SQK-RBK114.96) from Oxford Nanopore Technologies.

The final prepared library was then sequenced using the FLO-MIN114 MinION or GridION Flow Cell R10 version on the GridION Mk1 platform. The bases were uniquely aligned to the Genome Reference Consortium Human Build 38 (GRCh38) for the *CYP2C19* gene regions. Variant calling for small indels was performed using Clair3 static version 1.0.10. The following versions were used in the generation of this report:

Pharmcat:
PharmGKB database:

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TEST LIMITATIONS

- An individual having a variant in a gene not included in this test may change his/her predicted metabolizer profile or protein function. Additionally, this test cannot fully exclude the presence of pathogenic variants in genes that was not analyzed, genes with incomplete coverage, or regions not captured by our filtering strategies.
- This test does not account for extrinsic factors (such as lifestyle, drug-drug interactions, smoking status) or other intrinsic factors (renal or hepatic function, age, sex) that may affect an individual's ability to process medications. Thus, the clinician needs to consider the pharmacogenomic results in the context of patient's wider medical history.[3]

REFERENCE

Our main reference for clinical recommendation is the specific guidelines from CPIC and the European Society of Cardiology. Educational resource from the NHS UK was also used. Publication detail is as follows :

1. Scott SA, Sangkuhl K, Stein CM, Hulot JS, Mega JL, Roden DM, Klein TE, Sabatine MS, Johnson JA, Shuldiner AR. Clinical Pharmacogenetics Implementation Consortium guidelines for CYP2C19 genotype and clopidogrel therapy: 2013 update. Clinical Pharmacology & Therapeutics. 2013 Sep;94(3):317-23.
2. Magavern EF, Kaski JC, Turner RM, Drexel H, Janmohamed A, Scourfield A, Burrage D, Floyd CN, Adeyeye E, Tamargo J, Lewis BS. The role of pharmacogenomics in contemporary cardiovascular therapy: a position statement from the European Society of Cardiology Working Group on Cardiovascular Pharmacotherapy. European Heart Journal-Cardiovascular Pharmacotherapy. 2022 Jan;8(1):85-99.
3. Janmohamed A, Kulkarni S, Barker C, Newman B. Results: Patient with a known CYP2C19 genotype and coronary artery disease requiring clopidogrel [Internet]. NHS; 2024 [cited 2025 Jan 7]. Available from: https://www.genomicseducation.hee.nhs.uk/genotes/in-the-clinic/results-patient-with-a-known-cyp2c19-genotype-and-coronary-artery-disease-requiring-clopidogrel/

Technician :

Approved by :

Date of approval :