Kementerian Kesehatan
RS Cipto Mangunkusumo
Jalan Diponegoro Nomor 71
Jakarta 10430
1500135
www.rscm.co.id

RSCM CLINICAL RESEARCH UNIT (CRU) GENETIC TEST REPORT
METABOLIC HUB RESEARCH PROJECT

Patient Name	
Date of Birth	
Rekam Medis	
Gender	
Clinical Diagnosis	Familial Hypercholesterolemia (FH)
Symptoms	
Physician	dr. Dicky Tahapary, SpPD-KEMD., PhD
Genetic Counselor	dr. Widya Eka Nugraha, M.Si. Med.

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VARIANT GRADING

Pathogenic	variant that is certain to disrupt gene function or to be disease causing
Likely pathogenic	The variant may result in loss of gene function and thus can be considered the probable cause of the patient's disease.
Variant of unknown significance(VUS)	Variant of unknown biological significance -usually due to lack of knowledge
Likely Benign	Lower frequency variant with no reason to suspect a recessive or hypomorphic role, or likely neutral after functional/family studies
Benign	High frequency variant with no reason to suspect a recessive or hypomorphic role, or certainly neutral after functional family studies

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***Interpretation should be made by the physician who ordered the test or by the genetic counselor.**

Disclaimer & Methodology

- The interpretation of these genetic test results should be considered alongside clinical findings and cannot be used as the sole basis for clinical decisions. It is recommended to discuss the results with a genetic counselor or the healthcare professional who recommended the test.
- Test results should be evaluated in the context of clinical findings, including family history and other laboratory data.
- Rare polymorphisms may result in false positive or false negative outcomes.
- Genetic testing is generally highly accurate; however, inaccuracies can occur. These may be due to mislabeled samples, incorrect reporting of clinical or medical information, technical errors, or contamination of samples (for example, from prior blood transfusions or transplants), among other factors.
- Targeted sequencing was performed using GridION (Oxford Nanopore Technologies) with a coverage > 30X. The reference genome used was GRCh38.p14 (hg38). Variants were selected for analysis based on their relevance to familial hypercholesterolemia, focusing on the LDL-R gene. Analysis was conducted using the Epi2me software and Clair3, with further variant analysis and annotation supported by:

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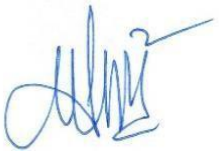
- SnpEff
- SnpSift
- ClinVar
- OMIM
- gnomAD
- dbSNP
- SIFT
- PolyPhen2

Genetic Counselor	Head of Clinical Research Unit RSCM	Laboratory Director of Clinical Research Unit RSCM
		
dr. Widya Eka Nugraha, M.Si. Med.	dr. Dicky Tahapary, SpPD-KEMD., PhD	dr. Selvi Nafisa Shahab, SpMK

PATIENT NAME:
DATE OF BIRTH: