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Kementerian Kesehatan
RS Cipto Mangunkusumo

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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.

VARIANT SUMMARY FOR FH

Gene/ Variant	Genotype	Assesment	Mode of Inheritance	Phenotype
None found	-	-	-	-

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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RESULTS INTERPRETATION

Gene/Variant	Interpretation of your genetic result
None found	We did not find any pathogenic or likely pathogenic variants associated with FH in your genetic profile. Familial hypercholesterolemia is a medical condition characterized by elevated serum cholesterol caused by genetic defects that can be passed to the patients' offsprings. The main genes that can cause the condition are LDL receptor (most common), apolipoprotein B (ApoB), or proprotein convertase subtilisin/Kexin type 9 (PCSK9).

*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: