

SPECIAL SG GENETIC DISEASES EVALUATION CENTER
MEDICAL ANALYSIS REPORT

LABORATORY LICENSE NO: GHDM-SM/34.39/01

Patient Name- Surname : Sample Collection Location :
Gender : Request Date :
Date of birth : Sample Collection Date :
TR ID Number : Sample Acceptance Date :
Protocol No. : Report Approval Date :
Sample No. : Report Publication Date / Report No. :
Sample Type :

Referring Center / Physician:

Reason for Reference:

Test Name: Hereditary Cancer Panel (59 Genes)

Method: Next Generation Sequence Analysis

Genes Examined: APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DDB2, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCC, FH, FLCN, GALNT12, HDAC2, HOXB13, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLE, POLH, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, SMAD4, STK11, TP53, TSC1, TSC2, VHL, WT1, XPA, XPC

Platform Used: Illumina Nextseq

Kit Used: SOPHiA GENETICS: The Custom Hereditary Cancer Solution (CHCS) Kit

Bioinformatics Analysis: SophiaDDM®

DATA ANALYSIS RESULT

59 Gene- SNV / INDEL Analysis:	CNV Analysis:
<p>NEGATIVE, Pathogenic/probably pathogenic¹No variant was detected.</p> <p>¹Variant class is determined according to ACMG criteria.</p>	<p>NEGATIVE, No deletion/duplication was detected in the examined genes.</p>

INTERPRETATION:

In the study conducted on the DNA material obtained from the patient's peripheral blood, no variant was observed that was definitely proven to cause the disease within the detection capacity of the methods used.

RECOMMENDATIONS:

**This section includes recommendations regarding the test performed and the results obtained.*

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QUALITY CONTROL RESULTS OF THE STUDY:

Average depth of target area:	X	At least 25X coverage of target area:	%
At least 50X coverage of target area:	%	At least 100X coverage of target area:	%

TECHNICAL SPECIFICATIONS AND RESTRICTIONS:

**Information is provided about the specification and limitations of the kit and technique used. This section also includes information about what is included or excluded from the report within the scope of the study and analysis.*

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REFERENCES:

-Richards, Sue, et al. "Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology." Genetics in medicine 17.5 (2015): 405.

- NCCN Guidelines Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic-Version 1.2023 — September 7, 2022

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Dr. Ceyhan Sayar, MD
Medical Genetics

Assoc. Prof. Dr. Kanay Yazarbaşı, MD
Medical Genetics

Private SG Genetic Diseases Evaluation Center has ISO 15189 Medical Laboratory Accreditation.

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Unsigned reports are invalid.

This result covers only the analyzed sample.

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Info: T:+90 212 276 01 66 M: +90 532 419 27 88 F: +90 212 276 01 56 www.sapiens.com.tr info@sapiens.com.tr

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