

SAPIENS SG GENETIC DISEASES DIAGNOSTIC CENTER
MEDICAL ANALYSIS REPORT

LABORATORY LICENSE NO: GHDM-SM/34.39/01

Patient Name : Sampling Location :
Gender : Test Request Date :
Date Of Birth : Sample Collection Date :
ID Number : Sample Acceptance Date :
Protocol Number : Report Approval Date :
Sample Number : Report Release Date/Report :
Number
Sample Type :

Referring Center / Physician :

Reason For Referral :

Test Name: Hereditary Breast Ovary Panel (BRCA 1-2)

Method: Next Generation Sequencing Analysis (NGS)

Used Platform: Illumina Miseq

Used Kits: Homologous Recombination Solution by SOPHiA GENETICS

Bioinformatics Analysis: Sophia DDM®

ANALYSIS RESULTS:

| |
|---|
| SNV / INDEL Analysis: |
| POSITIVE, The Tier IA c.5485dup (p.Glu1829Glyfs*51) variant was detected in <i>BRCA1</i> gene. No pathogenic variants detected in the <i>BRCA2</i> gene. |

INTERPRETATION:

| Tier IA | Tier IB | Tier IIC | Tier IID |
|--|----------------------------|----------------------------|----------------------------|
| <i>BRCA1</i> (NM_007294.4): c.5485dup (p.Glu1829Glyfs*51) | No variant was detected | No variant was detected | No variant was detected |

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In the study performed on the DNA material obtained from the patient's formalin fixed paraffin embedded sections, **Tier IA c.5485dup (p.Glu1829Glyfs*51)** pathogenic mutation of **BRCA1 (NM_007294.4)** was detected with 49.8% variant fraction.

BRCA-related cancers (Breast, Ovarian, Pancreatic Adenocarcinoma, Prostate; etc.) now recommend treatment with PARP (poly ADP-ribose polymerase) inhibitors for patients with germline or somatic **BRCA1/2** pathogenic/likely pathogenic variants, as PARP inhibitors have been demonstrated to be active in these patients. These agents include **niraparib, olaparib, and rucaparib** for chemotherapy-refractory ovarian cancer (NCCN Guidelines[®])².

RECOMMENDATIONS:

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

TECHNICAL SPECIFICATIONS AND LIMITATIONS:

**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:

1. 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.
2. Li, Marilyn M., et al. "Standards and guidelines for the interpretation and reporting of sequence variants in cancer: a joint consensus recommendation of the Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists." The Journal of molecular diagnostics 19.1 (2017): 4-23.
3. NCCN Guidelines Ovarian Cancer Continue Including Fallopian Tube Cancer and Primary Peritoneal Cancer Version 2.2023 — June 2, 2023
4. NCCN Guidelines Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic Version 2.2024 — September 27, 2023

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Özel 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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