

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: BRCA MASTR Dx Kit, Multiplicom

Bioinformatics Analysis: Sophia DDM®

ANALYSIS RESULTS:

SNV / INDEL Analysis:

POSITIVE,

Heterozygous for *BRCA1* (NM_007294.4) c.5137del (p.Val1713*) pathogenic variant was detected.

No pathogenic variants detected in the *BRCA2* gene.

INTERPRETATION:

Heterozygous pathogenic c.5137del (p.Val1713*) variant was detected in *BRCA1* gene.

The c.5137del (p.Val1713*) variant is a deletion that causes a stop codon at position 1713 in the protein. Loss-of-function variants in *BRCA1* are known to be pathogenic (PMID: 20104584). ClinVar database classifies this variant as 'Pathogenic' related to Breast-ovarian cancer, familial, susceptibility to, 1 syndrome (ClinVar ID: 55411). dbSNP database classifies this variant as 'Pathogenic' (dbSNP: rs80357997). This variant is present with low frequency in population databases (gnomAD: 0.00000398). According to ACMG criterias¹, this variant is classified as 'pathogenic'.

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The patient has an elevated/high lifetime risk of developing further *BRCA1*-related cancers.

The *BRCA1* gene encodes a protein that plays an important role in DNA repair, control of cell cycle checkpoints, and maintenance of genomic stability. This protein is a tumor suppressor protein that plays a role in forming the *BRCA1*-related genome surveillance complex (BASC), together with other tumor suppressors, DNA damage sensor and signal transmitters. The germline pathogenic variants in the *BRCA1* gene cause more than 60% risk for breast cancer, 7–26% risk for prostate cancer and 39-58% risk for ovarian cancer².

Variant Table:

| Gene (Reference Sequence) | Variant | Classification [¶] | Status |
|-------------------------------|---------|-----------------------------|--------|
| <i>BRCA1</i> (NM_007294.4) | | | |

[¶] Variant class is determined based on American College of Medical Genetics and Genomics (ACMG) criterias.

RECOMMENDATIONS:

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

QUALITY CONTROL RESULTS

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| | | | |
|---------------------------------------|---|--|---|
| Target region's average coverage: | x | Target region's minimal 25X coverage: | % |
| Target region's minimal 50X coverage: | % | Target region's minimal 100X coverage: | % |

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.*

REFERENCES:

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Referring Center / Physician :

Reason For Referral :

1. Richards S 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(PMID: 25741868)
2. Genetic/Familial High Risk Assessment: Breast, Ovarian, and Pancreatic NCCN Guideline Version 2.2024 — September 27, 2023
3. <https://portal.biobase-international.com/hgmd/pro/gene.php?gene=BRCA1>
4. <https://portal.biobase-international.com/hgmd/pro/gene.php?gene=BRCA2>
5. Luca Livraghi and Judy E. Garber. BMC Med. 2015; 13(1):188 (PMID: [26268938](https://pubmed.ncbi.nlm.nih.gov/26268938/)).
6. Goff BA1., J Gynecol Oncol. 2013 Jan;24(1):83-91. doi: 10.3802/jgo.2013.24.1.83. Epub 2013 Jan 8.
7. <https://brcaexchange.org/variants>

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

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This result covers only the analyzed sample.

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Verification Link:

