BRCAssure® Comprehensive *BRCA1* and *BRCA2* Analysis

labcorp

Specimen ID:
Control ID: **Lab Case ID:** **Acct #:** **Phone:**

REPORT, SAMPLE

Patient Details	Specimen Details	Physician Details
DOB:	**Date collected:**	**Ordering:**
Age(y/m/d):	**Date received:**	**Referring:**
Gender: F	**Date entered:**	**ID:**
Patient ID:	**Date reported:**	**NPI:**

Specimen Type: Whole Blood
Clinical Indication: Personal and/or family history of Hereditary Breast and Ovarian Cancer (HBOC)

RESULTS

NEGATIVE FOR PATHOGENIC VARIANTS

No clinically significant variants were identified.
At least one variant of uncertain significance (VUS) was identified; see Additional Clinical Information for details.

GENE	VARIANT
BRCA1	**NEGATIVE** No pathogenic variants were identified.
BRCA2	**NEGATIVE** No pathogenic variants were identified.

INTERPRETATION

No pathogenic variants were identified.

ADDITIONAL CLINICAL INFORMATION

Variant(s) of uncertain significance (VUS) in this individual

BRCA2	Heterozygous for c.4354C>G (p.Gln1452Glu) (VUS - uncertain significance). Insufficient information is available to classify this variant as either benign or pathogenic.

The significance of VUS in association with relative risk for HBOC (Hereditary Breast and Ovarian Cancer Syndrome) is uncertain at the time of reporting. Evidence for variant classification is available upon request.

SAMPLE

Electronically released under the direction of Toni Prezant, PhD, FACMG

Testing Performed at Laboratory Corporation of America Holdings, 1912 T.W. Alexander Drive, RTP, NC 27709-0150, 1-800-345-4363, Medical Director: Anjen Chenn, M.D., Ph.D.

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ADDITIONAL CLINICAL INFORMATION

NCCN Guidelines

When BRCA1 and BRCA2 results are negative, additional testing may be helpful for some patients. Guidelines from the National Comprehensive Cancer Network® (NCCN®) recommend considering germline genetic testing for additional breast, ovarian, prostate, and/or pancreatic cancer susceptibility genes in patients meeting any of the criteria in the table below. To discuss comprehensive genetic testing, a Labcorp Genetic Coordinator is available at 800-345-4363.

Breast cancer diagnosed ≤ age 50
Ovarian cancer, pancreatic cancer, male breast cancer, or metastatic/high-risk prostate cancer at any age
Breast cancer diagnosed at any age and one of the following: • To aid in PARP inhibitor or olaparib treatment • Triple negative breast cancer • Multiple primary breast cancers • ≥ 3 total diagnoses of breast cancer in patient and/or close blood relatives
Breast or prostate cancer diagnosed at any age and one of the following: • ≥ 1 close relative with breast cancer ≤ age 50 or with triple negative breast cancer at any age • ≥ 1 close relative with ovarian, pancreatic, male breast, or metastatic/high-risk prostate cancer • ≥ 2 close relatives with breast or prostate cancer at any age • Ashkenazi Jewish ancestry
Patients with a first or second degree relative meeting certain criteria in this table may consider germline genetic testing as well. Complete criteria may be found at NCCN.org.

RECOMMENDATIONS

Based on information provided, this patient meets National Comprehensive Cancer Network® (NCCN®) clinical guidelines that recommend considering testing for additional genes. See Additional Clinical Information for more details. Labcorp Genetic Coordinators are available at (800) 345-GENE to discuss additional genetic testing if desired.

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of these results, as well as recommendations for testing family members. To access Labcorp Genetic Counselors please visit https://womenshealth.labcorp.com/genetic-counseling or call (855) GC-CALLS (855-422-2557).

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METHODS AND LIMITATIONS

Next-generation sequencing: Genomic regions of interest are selected using a custom capture reagent for target enrichment and sequenced via the Illumina(R) next generation sequencing platform. Regions of interest include all exons and intron/exon junctions (+/-20 nucleotides) of the *BRCA1* (NM_007294.3) and *BRCA2* (NM_000059.3) genes. Regions of interest may be extended to include intronic sequences known to harbor pathogenic/likely pathogenic variants. Sequencing reads are aligned with the human genome reference GRCh37/hg19 build. Minimum mean coverage is 40X. Any segment failing minimum read depth coverage is rescued by bi-directional Sanger sequencing to complete sequence analysis. Variants, including SNVs and CNVs, are identified using a custom bioinformatics pipeline.

Reported variants: Pathogenic and likely pathogenic variants and variants of uncertain significance (VUS) are reported. Non-deletion variants are specified using the numbering and nomenclature recommended by the Human Genome Variation Society (HGVS, http://www.hgvs.org/). Benign variants are not reported. Variant classification and confirmation are consistent with ACMG standards and guidelines (Richards, PMID:25741868; Rehm, PMID:23887774). Detailed variant classification information is available upon request. A variant of uncertain significance (VUS) should not be used in clinical decision making; a VUS is classified based on inadequate or conflicting evidence regarding its pathogenicity or clinical relevance.

Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations, including rearrangements, gene fusions, or variants in regions or genes not included in this test, or possible inter/ intragenic interactions between variants. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: genetic variants, pseudogene interference, technical handling, blood transfusions, bone marrow transplantation, mislabeling of samples, or erroneous representation of family relationships. For heterozygous variants in the same gene the assay cannot determine whether they are on the same or different chromosome; to determine phase and clinical significance, rarely, parental testing may be required. Exact breakpoints of exon-level deletions/duplications are not determined. The presence of an inherited cancer syndrome due to a different genetic cause cannot be ruled out. Any interpretation should be clinically correlated with information about the patient's presentation and relevant family history.

REFERENCES

1. NCCN Genetic/Familial High Risk Assessment: Breast, Ovarian, and Pancreatic. Version 2.2023.
2. Petrucelli, et al. *BRCA1*- and *BRCA2*-Associated Hereditary Breast and Ovarian Cancer. GeneReviews, updated 2022. PMID: 20301425.

This test was developed and its performance characteristics determined by Labcorp. It has not been cleared or approved by the Food and Drug Administration.

PERFORMING LABS

Component Type	Performed At	Laboratory Director
Technical component, processing	Laboratory Corporation of America, 1912 TW Alexander Drive, RTP, NC, 27709-0150	Anjen Chenn, MD, PhD
Technical component, analysis	Laboratory Corporation of America, 1912 TW Alexander Drive, RTP, NC, 27709-0150	Anjen Chenn, MD, PhD
Professional component	Laboratory Corporation of America, 6401 Pat Avenue, West Hills, CA, 91307	Toni Prezant, PhD, FACMG

For inquiries, the physician may contact the lab at 800-345-4363

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