Reporting Title: BRCA1/BRCA2 Testing

Test Definition: BRCA
Testing Location: Tucker, GA
Reporting Location: Melville, NY

Description:
Pathogenic variants in the BRCA1 and BRCA2 genes cause hereditary breast and ovarian cancer syndrome (HBOC), an autosomal dominant cancer predisposition syndrome. Individuals with pathogenic variants in these genes are at a significantly increased risk for breast, ovarian and other cancers.1-3

Analytical Method(s):
See Attached Example Reports

Patient Preparation:
Counsel patient on BRCA screening

Specimen Requirements:
Container/Tube: Lavender-top Vacutainer® tube (EDTA).
Specimen Volume: 1.0 ml of spun plasma or 5 ml of unspun whole blood.
Specimen Stability: Serum samples are stable at ambient temperature for 6 days.
Specimen Rejection Criteria: hemolysis, lipemia, incorrect tube type
Specimen Collection Instructions See also Blood Specimen Collection from Venipuncture Instruction Manual.

Specimen Shipping Requirements:

<table>
<thead>
<tr>
<th>Test Type</th>
<th>Sample Type</th>
<th>Tube Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRCA1 and BRCA2 Hereditary</td>
<td>Whole Blood</td>
<td>EDTA (purple top)</td>
</tr>
<tr>
<td>Cancer Screening</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genetic Carrier Screening</td>
<td>Whole Blood</td>
<td>EDTA (purple top)</td>
</tr>
</tbody>
</table>

Collection Instructions

- For each test, collect 5-10 mL whole blood and ship in the original collection tube without centrifugation place collection tube inside the outer screw-top tube to avoid potential for breakage or leakage.
- Can be shipped at ambient or refrigerated temperature Monday through Friday.
- Specimens must be received at NTD Labs within 4 days of collection.
- Do not freeze.

CAUSES FOR REJECTION: Whole blood frozen, specimens beyond their acceptable length of time from collection as listed, received in trap containers or specimen types other than those listed.

Shipping Instructions

- Label all specimen tubes with two unique patient identifiers (specimen labels are available for NTD’s Genetic Testing Requisition Form).
- Place tube into provided shipping canister and close tightly. Place closed shipping canister into bag. Place the biohazard bag into the collection kit box along with the completed test requisition form.
- Ship specimens within 24 hours. Refrigerate specimens that will be delayed more than 24 hours before shipping. Friday shipments must be labeled for Saturday delivery. All specimens must be labeled with name and date of birth or name and MRN. A test requisition form must accompany each specimen.

Ship specimens FedEx Priority Overnight to: Eurofins NTD, LLC.
80 Ruland Road, Suite 1
Melville, NY 11747
Additional Information: N/A

CPT Codes:

MM070 – 81162  
MM071- 81162  
MM072- 81213, 81228

Reference Values:

An Interpretive Report will be provided

Supplemental Report:

No

Testing Algorithm:

Follow-up testing:
  1. Mutation Identified: Genetic counseling
  2. Variants of unknown significance detected by Next Gen Sequencing may require further studies of the patient and/or family members.

Consents/Authorizations:

  1. A patient courtesy consent form is available for Genetic Testing.
  2. A New York State informed consent acknowledgment form is required for genetic Testing. This is to be signed by the healthcare provider and kept on file at Eurofins NTD.
  3. Patient consent for research.

Disclaimers:

See Attached Example Reports

Test Requisition Instructions

Complete Physician Information, Patient Information and Clinical Information sections. Be sure to check off the specific Test Request.

Specimen Labels – Preprinted with the requisition number. Please enter the patients last and first names EXACTLY as they appear on the requisition form. Affix the label to the patient specimen. Please complete the date drawn and drawn-by fields.

Billing Information – Provide a photocopy of the front and back of insurance card or print the information in the required fields. Please provide credit card information to cover tests ordered requiring additional charges.
Test Requisition Form

Physician Information

Ordering Physician
Ordering Physician Signature (Required for Medicare Patients)
Referring OBGYN
Referring OBGYN Phone

Specimen Labeling

Sample Type: Whole Blood in EDTA Tube Required
Data Drawn ______/______/______
Time Drawn ______:____ AM or PM

Enter patient’s name on specimen identification label EXACTLY as it appears on the Requisition Form below. Peel off label and affix to the specimen tube. Two forms of patient’s ID MUST appear on both the Test Requisition Form and the specimen tube.

Patient Information

Last Name
First Name
Date of Birth ______/______/______
Address
City
State
Zip
Sex: F M Unknown Ambiguous
E-mail

Test Request

☐ MM470 Pan-Ethnic Carrier Screen; Gene Sequencing Panel
☐ MM460 Pan-Ethnic Carrier Screen; Targeted Mutation Panel
☐ MM600 ACOG/ACMG Carrier Screen; Targeted Mutation Panel
☐ Sialidase-Abnormal
☐ MM500 Spinach Muscular Atrophy; Carrier Screen
Included in MM470, MM460, and MM500
☐ MPRAK Fragile X; CGG Repeat Analysis
Included in MM460 and MM460
☐ MM500 CF Transmembrane Conductivity Defect Panel
Included in MM460, MM460, and MM500
BRCA Screening Test Options
Hereditary Breast and Ovarian Cancer Syndrome
☐ MM070 BRCA1/BRCA2 Gene Sequencing and Deletion/Duplication Panel
☐ MM060 BRCA1/BRCA2 Gene Sequencing Panel
☐ MM070 BRCA1/BRCA2 Deletion/Duplication Panel
Qualifying analysis is required before deletion/duplication analysis by targeted DH array. If sequencing is performed outside of NTD Genetics, please submit a copy of the sequencing report with the test requisition.

Ethnicity

☐ African American
☐ Asian
☐ European/Caucasian
☐ Finnish
☐ E. Indian
☐ Jewish-Sephardic
☐ Mediterranean
☐ Native American
☐ Hispanic
☐ Ashkenazi Jewish
☐ Adopted
☐ Other

Please Check All

☐ Blood transfusion past 90 days
☐ Bone marrow transplant
☐ Pregnant or family member pregnant. Due to ethical considerations, we cannot provide this test.
☐ Results will directly impact patient treatment
☐ Family History of genetic disease
☐ If there is a partner’s sample, partner’s name

Please complete the patient information on the back of this form. Please provide a pen and paper to write the information.

Billing Information

Please attach a copy of the front and back of the patient’s insurance card or provide information below.

Insurance Company
Plan Name
Subscriber’s Last Name, First Name
Insurance ID
Insurance Claim Address
Secondary Insurance Information

Insured by: Eurofins NTD, LLC, to obtain and release all medical and other information and to directly bill and submit claims to Medicare, Medicaid, Medicare Supplemental and/or insurance providers. Certain Eurofins NTD, LLC, provides to me. I assign insurance benefits to Eurofins NTD, LLC, and acknowledge that charges that are not covered by insurance, including any applicable co-payments, deductibles, co-insurance, or non-authorized care or services, are my responsibility, and I agree to pay for such charges.

Patient Signature (required for all tests)

EGL Lab Use Only
Unboxed by
Received
NTD Lab Use Only
Specimen Received:
☐ Whole Blood in EDTA

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NTD Genetic Testing

I understand no tests other than those authorized shall be performed on my sample and that the sample shall be discarded within sixty (60) days as specified in the consent below. Eurofins NTD, LLC. performs research to help develop and provide safe and effective screening tests and to improve biomedical knowledge. Research and development studies are conducted to improve existing tests and validate new tests. My permission to identify my sample in research and development studies is entirely voluntary.

I give permission for EGL Genetics Diagnostics to release the remaining biological sample to Eurofins NTD, LLC. to retain for future research.

Patient or Legal Guardian Signature: __________________________ Date: __________/________/______

Instructions: The accurate interpretation and reporting of genetic test results is contingent upon the clinical information provided and best possible service, please check the applicable clinical information below.

### Clinical Information

<table>
<thead>
<tr>
<th>Perinatal History</th>
<th>Behavior/Psychiatric</th>
<th>Cardiac</th>
<th>Hearing/Vision</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prematurity</td>
<td>Autism</td>
<td>Atrial septal defect</td>
<td>Hearing loss</td>
</tr>
<tr>
<td>Intratable growth restriction</td>
<td>Pervasive developmental delay</td>
<td>Ventricular septal defect</td>
<td>Specifying</td>
</tr>
<tr>
<td>Obesity</td>
<td>Attention deficit hyponatremia disorder</td>
<td>Coarctation of the aorta</td>
<td>Specifying</td>
</tr>
<tr>
<td>Polycythemia</td>
<td>Anxiety</td>
<td>Tetralogy of Fallot</td>
<td>Specifying</td>
</tr>
<tr>
<td>Polycythemia</td>
<td>Behavioral/psychiatric abnormality</td>
<td>Other structural heart defect</td>
<td>Specifying</td>
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<td>Polycythemia</td>
<td>Other</td>
<td>Other cardiac abnormality</td>
<td>Specifying</td>
</tr>
<tr>
<td>Non-Immune hydronephrosis</td>
<td>Other</td>
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<td>Other</td>
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<td>Specifying</td>
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<th>Cutenous</th>
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<th>Microphthalmologic</th>
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<td>Hyperpigmentation</td>
<td>Seizures</td>
<td>Dysmorphic facial features</td>
<td>Contractures</td>
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<td>Hypopigmentation</td>
<td>Hypotonia</td>
<td>Ear malformation</td>
<td>Club foot</td>
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<td></td>
<td>Hyperpnea</td>
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<td>Diaphragmatic hernia</td>
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<td>Hypoventilation</td>
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<td>Limb anomaly</td>
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<td>Other</td>
<td>Cystic hygroma</td>
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<td>Other</td>
<td>Other</td>
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</tr>
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### HEREDITARY CANCER TEST REQUISITION FORM

#### Clinical Information (Pedigree may be submitted on a separate page)

<table>
<thead>
<tr>
<th>Cancer Type</th>
<th>Personal History</th>
<th>Family History</th>
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<tbody>
<tr>
<td>Breast</td>
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<tr>
<td>Ovarian</td>
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<td>Colon</td>
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<td>Polyps</td>
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<td>Endometrial</td>
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<td>Sarcoma</td>
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<tr>
<td>Lung</td>
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<th>Question ID</th>
<th>Description</th>
<th>Type</th>
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<td>Ethnicity</td>
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<td>- European/Caucasian</td>
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<td></td>
<td>- E. Indian</td>
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<td>- Jewish-Sephardic</td>
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<td>- Native American</td>
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<td>- Ashkenazi Jewish</td>
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<td>MM070 Test Panel (BRCA1/BRCA2 Gene Sequencing and Dup/Del Panel)</td>
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<td>- Yes</td>
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<td></td>
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<td>- No</td>
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<td>Answer List</td>
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<td>- Yes</td>
<td></td>
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<td>- No</td>
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<tr>
<td>BRCA</td>
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<td>MM072 Test Panel (BRCA1/BRCA2 Dup/Del Panel)</td>
<td>Answer List</td>
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<tr>
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## EMR Result Codes:

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<th>Data Type</th>
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<th>LOINC</th>
<th>Name</th>
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<th>Comments</th>
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<tr>
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<td>BRCA1/BRCA2 Gene Sequencing and Dup/Del Panel</td>
<td>Yes</td>
<td>See PDF for Results</td>
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<td>CE</td>
<td>MM071</td>
<td>59041-4</td>
<td>BRCA1/BRCA2 Gene Sequencing Panel</td>
<td>Yes</td>
<td>See PDF for Results</td>
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<td>CE</td>
<td>MM072</td>
<td>59041-4</td>
<td>BRCA1/BRCA2 Dup/Del Panel</td>
<td>Yes</td>
<td>See PDF for Results</td>
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<td>DGD</td>
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<td>Included if demographic data is available (contained in NTEs)</td>
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<td>Footer</td>
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<td>Only Displayed if footer is</td>
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<td>Notification</td>
<td>Yes</td>
<td>Included for Unsatisfactory Specimens Only</td>
<td>available</td>
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</table>

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Example Report

Hereditary Breast and Ovarian Cancer Syndrome:
BRCA1 and BRCA2 Gene Sequencing and Deletion/Duplication

Results: Pathogenic variant detected. One copy of a c.5035_5039delCTAAT pathogenic variant was detected in the BRCA1 gene of this individual.

Interpretation

A sample from this individual was referred to our laboratory for molecular testing for hereditary breast and ovarian cancer syndrome (HBOC). HBOC is an autosomal dominant cancer predisposition syndrome that signifies the risk for breast, ovarian, and other cancers. Pathogenic variants in the BRCA1 and BRCA2 genes cause single pathogenic variant in one copy of either the BRCA1 or BRCA2 gene is associated with disease.

Sequence analysis of the coding regions of the BRCA1 and BRCA2 genes detected the following:

<table>
<thead>
<tr>
<th>Gene</th>
<th>Exon/Intron</th>
<th>Nucleotide change</th>
<th>Zygosity</th>
<th>Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRCA1</td>
<td>Ex16</td>
<td>c.5035_5039delCTAAT</td>
<td>Heterozygous</td>
<td>Pathogenic</td>
</tr>
</tbody>
</table>

The c.5035_5039delCTAAT variant has been reported in individuals with hereditary breast cancer and is expected to cause disease.

These results must be interpreted in the context of this individual’s clinical and biochemical profile. Gene is recommended. For more information, please visit geneticleab.emory.edu or call 855-855-74477 (toll-free) laboratory genetic counselor or consult with a laboratory director.

Notes: A list of benign variants unrelated to disease identified in this individual is available upon request evaluated by their reported frequency. Variants that have a population frequency greater than expected prevalence of the disease in the general population are considered to be benign variants. Silent variants are unless known to be pathogenic or other evidence suggests potential disruption of splicing. The interpretation is based on our current understanding of the BRCA1 and BRCA2 genes. These interpretations may change more information about the genes becomes available. Visit EmVClass (geneticleab.emory.edu) for curers of variants. Possible diagnostic errors include sample mix-ups, genetic variants that interfere with analysis.
able to be analyzed. Nucleotide numbering is based on GenBank accession numbers NM_007294.3 and 1 nucleotide 1 corresponds to the A of the start codon ATG.

This sample was analyzed using a comparative genomic hybridization (CGH) array custom designed for BRCA1 and BRCA2 genes. Rarely, probe coverage may be limited or absent in some exons due to the target sequence.

References:
1 Musolino (2005) Tumori 91: 505
2 www.ncbi.nlm.nih.gov/snp,
3 evs.gs.washington.edu/EVS/,
4 browser.1000genomes.org/,
5 exac.broadinstitute.org/
6 gnomad.broadinstitute.org/

Testing was performed and results originated from EGL Genetic Diagnostics LLC (CLIA#: 11D0683478; CAP#: 7181693, Lora J J FACMG, Director), 2480 Mountain Industrial Boulevard, Tucker, GA 30084. Pursuant to the requirements of CLIA ’88, this test was its performance validated by EGL Genetic Diagnostics LLC. It has not been cleared or approved by the U.S. Food and Drug Admin. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.

Medical Consultant, Laboratory Director, Molecular Genetics
EGL Genetic Diagnostics, LLC
EGL Genetic Diagnostics, LLC

This case has been reviewed and electronically signed by a Laboratory Director.
Physician Attestation to Obtain Informed Consent

Acknowledgement of Practitioner’s Obligation to Obtain Informed Consent for Genetic Testing

- I understand that the New York State Department of Health, Clinical Laboratory Standards Practice requires NY-permitted laboratories to notify ordering practitioners that they aim to obtain informed consent for genetic testing from their patients.
- I am aware that Eurofins, NTD, LLC (PFI 3173) (“Eurofins”) provides a genetic test consent that includes test-specific information to aid me in fulfilling my obligations to obtain informed consent.
- I understand that informed consent for the NIPT tests must be obtained utilizing the form for that test, copies of which have been provided to me by Eurofins.
- I have been apprised that I may obtain these forms in hard copy from Eurofins, and they are also available on the Eurofins website (http://www.ntdlabs.com/providers/consent_forms.php).
- I acknowledge that this information has been made available to me for patient use in making the informed consent process.
- I acknowledge that I have reviewed and understand the informed consent for genetic testing that I am ordering as described above and that the information on the form shall be conveyed to each patient and/or their guardian in obtaining full and effective informed consent for each genetic test I order.
- I confirm that I shall maintain documentation that I obtained informed consent for each of my patients’ medical charts.
- I confirm that I will make available and provide a copy of the signed patient consent for Eurofins NTD, LLC upon request.

__________________________________    __________________________
Practitioner Signature                     Date

__________________________________
Printed Name

__________________________________
Hospital, Facility or Clinic Name

__________________________________
Address

__________________________________
City, State, Zip

__________________________________
Phone #
Patient Informed Consent

Patient Informed Consent for Hereditary Breast and Ovarian Canc

PLEASE NOTE: A signed Physician Acknowledgement of NY Informed Consent for Gene be on file at Eurofins NTD, LLC to permit testing and processing.

Test Specific Information (check those that apply)

- **MM071 BRCA1/BRCA2 Gene Sequencing Panel**
  - In-solution hybridization of all coding exons is performed on the patient’s genomic DNA. Direct sequencing of the cap performed using next generation sequencing. The patient’s gene sequences are then compared to a standard reference. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further investigation.
  - Limitations:
    - Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis.
    - Large deletions/duplications will not be detected by this analysis.
    - Results of molecular analysis should be interpreted in the context of the patient’s clinical/biochemical phenotype.
    - Clinical sensitivity is unknown. Analytical sensitivity is approximately 99%.

- **MM072 BRCA1/BRCA2 Deletion/Duplication Panel**
  - DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH overlapping probes which cover the entire genomic region.
  - Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed for Genetics, please submit a copy of the sequencing report with the test requisition.
  - Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Off-target copy number variants may be identified that may or may not be related to the gene.
  - Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown significance will not be reported.
  - Limitations:
    - Detection is limited to duplications and deletions. The CGH array will not detect point or structural mutations.
    - Results of molecular analysis must be interpreted in the context of the patient’s clinical and/or biochemical phenotype.

- **MM070 BRCA1/BRCA2 Gene Sequencing and Deletion/Duplication Panel**
  - Both BRCA1/BRCA2 gene sequencing and deletion/duplication panels described above are performed.

General Information, Limitations, and Risks:

- Mutations in the genes BRCA1 and BRCA2 cause hereditary breast and ovarian cancer syndrome (HBOC), an autosomal recessive syndrome. Mutations in these genes are rare and account for only a small percentage of cancers; about 10% of ovarian cancers are due to mutations in the BRCA1 or BRCA2 genes. Individuals with mutations, however, are at a significantly increased risk for developing breast, ovarian, and other cancers than those in the general population.

This analysis can have the following outcomes:

- Positive: A pathogenic variant (disease-causing) could be identified in one or more of the genes being tested for and identified as being affected.
- Negative: No pathogenic variant is identified. This reduces the risk of being affected by the diseases specifically tested for.
- Inconclusive: Due to technical issues, the results were inconclusive and the test might need repeating. Results may not be complete due to the identification of a variant of unknown significance.
Patient Informed Consent for Hereditary Breast and Ovarian Canc

to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA or to the complexity of the testing itself. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist

• This DNA test requires a blood sample which has risks associated with obtaining the sample. Additional samples may be damaged in shipment or inaccurately submitted.

• It is the responsibility of the referring physician or health care provider to understand the specific use and limitations of the test and to educate the patient regarding these limitations. Additional information describing indications, methodology and interpretation is found on the EGL website at: https://www.egl-eurofins.com/

• Accurate interpretation of test results is dependent upon the patient’s clinical diagnosis or family medical history and on the understanding of the laboratory results. Genetic testing in family members can sometimes reveal that true biological relationships are not consistent with the reported clinical relationships. For example, non-paternity may be revealed, which means that the father of an individual is not the true biological father.

• It is the responsibility of the referring physician or health care provider to understand the specific use and limitations of the test and to educate the patient regarding these limitations.

• Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the referring physician, who will then review and discuss the test results with me.

• Patient-identifying results and information at Eurofins NTD, LLC will remain confidential and may only be released to or disclosed to other physicians or permitted or required by applicable law.

• I understand no tests other than those authorized shall be performed on my sample and that the sample shall be destroyed 7 days after being taken. However, Eurofins NTD, LLC performs research and development studies to improve and to validate new tests and to advance biomedical knowledge. I and my heirs will not receive payments, benefits, or rights to any research discoveries. Patient permission is requested for the use of patient-de-identified sample in research and development at voluntary.

• If I have additional questions, I understand that I may wish to obtain further professional genetic counseling prior to co-testing.

My signature below acknowledges my voluntary participation in DNA-based testing ordered by my physician for Hereditary Ovarian Cancer Syndrome (HBOC) in an attempt to determine whether I am at increased risk to be affected by the condition. It verifies that I have been appropriately counseled about the testing process and the different possible outcomes.

Patient/Guardian Signature: ____________________________

Printed Name: ____________________________

Date: ____________________________

Physician/Counselor/ Clinician Statement:

I have explained Hereditary Breast and Ovarian Cancer Syndrome (HBOC) testing to the patient/parent/guardian. The cellular limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test counseling. I will use my independent professional judgment and the patient’s best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device attention by Eurofins NTD, LLC or others.

Healthcare Provider Signature: ____________________________

Printed Name: ____________________________

Date: ____________________________
Physician Information Brochure

Hereditary Cancer Screening
Address a Broader Range of Women’s Health Needs

Pathogenic variants in the BRCA1 and BRCA2 genes cause hereditary breast and ovarian cancer syndrome (HBOC), an autosomal dominant cancer predisposition syndrome. Individuals with pathogenic variants in these genes have an increased risk of breast, ovarian, and other cancers.1-3

BRCA1 pathogenic variant
50–80% risk of developing breast cancer
Up to 44% risk of developing ovarian cancer

BRCA2 pathogenic variant
40–70% risk of developing breast cancer
Up to 27% risk of developing ovarian cancer

Guidelines recommend BRCA screening for at-risk patients
This includes those with a personal or family history of:

- Early-onset breast cancer (<50 years of age), bilateral breast cancer or triple negative (PR/ER/HER2 negative) breast cancer (<60 years of age)
- Two primary breast cancers or a diagnosis of both breast and ovarian cancer in one individual
- Personal or family history of male breast cancer
- Ovarian cancer at any age
- Ethnicity with a higher mutation frequency (e.g., Ashkenazi Jewish)

NTD Eurofins offers three testing options:
- Hereditary Breast and Ovarian Cancer Syndrome: BRCA1/BRCA2 Gene Sequencing and Deletion/Duplication Panel
## Comprehensive BRCA Screening for Your At-Risk Patients

<table>
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<th>Panel</th>
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| (MM070) Hereditary Breast and Ovarian Cancer Syndrome: BRCA1/BRCA2 Gene Sequencing and Deletion/Duplication Panel™ | - Turnaround time: 3 weeks  
- Hereditary Breast  
The most comprehensive screening option that uses next-generation sequencing (NGS) of captured gene-targeted comparative genomic hybridization array for deletion and duplication analysis. These methods will detect 90% of the known pathogenic related to HBOC. |
| (MM071) Hereditary Breast and Ovarian Cancer Syndrome: BRCA1/BRCA2 Gene Sequencing Panel* | - Turnaround time: 3 weeks  
- Analytical Sensitivity: 99%  
Direct sequencing of the captured regions is performed using NGS. This method detects 90% of known pathogenic variants related to HBOC. |
| (MM072) Hereditary Breast and Ovarian Cancer Syndrome: BRCA1/BRCA2 Deletion/Duplication Panel* | - Turnaround time: 7 days  
- Analytical Sensitivity: 99%  
DNA isolated from peripheral blood is hybridized a gene-targeted CGH array to detect deletions or duplications. This method detects 10% of known pathogenic variants related to HBOC. |

*Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign or variants of unknown significance. Variants of unknown significance may require additional genetic studies of the patient and/or family members.  
*Only known pathogenic off-target copy number variants will be reported.

## The NTD Eurofins Advantage
- Leader in high-quality prenatal testing for over 30 years.  
- Supported by the expertise of Eurofins’ EGL Genetics, providing cutting-edge testing that spans from common genetic disorders, cytogenetics to molecular and biochemical testing.  
- Personalized customer support with direct access to lab directors and genetic counseling support.
Patient Information Brochure

Who is at risk for a BRCA pathogenic variant?

You may be at increased risk if you have a personal or family history of:

- Early-onset breast cancer (<50 years of age), bilateral breast cancer or triple negative breast cancer (<60 years of age)
- Two primary breast cancers or a diagnosis of both breast and ovarian cancer
- Personal or family history of male breast cancer
- Ovarian cancer at any age
- Ethnicity with a higher pathogenic variant frequency (e.g., Ashkenaz Jewish)

Should I be tested?

A review of your family history is important when considering BRCA testing. Medical guidelines support testing for people with a high risk or strong family history of breast or ovarian cancer.

Why NTD Eurofins?

NTD has led in the research and development of prenatal screening protocols and tests for more than 20 years. Today, we serve universities, medical centers, hospitals, laboratories, obstetricians and maternal fetal medicine specialists around the world—providing women’s health screening services that help healthcare providers and patients make more informed medical decisions.

Supported by the expertise of Eurofins’ EOL Genetics, providing cutting-edge testing that spans from common to rare genetic diseases, heterogeneous to molecular and biochemical testing.

For more information about NTD BRCA testing, please speak with your healthcare provider, or call us at 1-888-NTD-LABS (683-5227).

References:


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Know your risk, know your options

While most cases of cancer happen by chance, up to 10% of cases are due to a change in a gene, or pathogenic variant, that has been passed down from generation to generation. This is called hereditary cancer.

What is BRCA testing?
The BRCA1 and BRCA2 (breast cancer 1 and 2) genes are found in everyone's normal genetic material. But people with pathogenic variant in these genes are at a higher risk of developing breast, ovarian or other cancers.

BRCA1 pathogenic variant
50–80% risk of developing breast cancer
Up to 44% risk of developing ovarian cancer

BRCA2 pathogenic variant
40–70% risk of developing breast cancer
Up to 27% risk of developing ovarian cancer

Genetic testing for BRCA1 and BRCA2 pathogenic variant—through a blood sample—can help provide further information about your risk for developing cancer.

Which tests does NTD offer?
NTD offers three BRCA testing options:

- Hereditary Breast and Ovarian Cancer Syndrome:
  BRCA1/BRCA2 Gene Sequencing and Deletion/Duplication Panel
- Hereditary Breast and Ovarian Cancer Syndrome:
  BRCA1/BRCA2 Gene Sequencing Panel
- Hereditary Breast and Ovarian Cancer Syndrome:
  BRCA1/BRCA2 Deletion/Duplication Panel

What do they mean?

Positive result
A positive test result means you have a pathogenic variant of BRCA and have an increased risk of developing breast or ovarian cancer. It is important to discuss the implications of this test result with your doctor.

Negative result
A negative test result means you do not carry a pathogenic variant of BRCA and do not have an increased risk of developing breast or ovarian cancer.

Uncertain result
An uncertain result means the test was inconclusive and additional testing may be recommended.

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