BRCA1 and BRCA2 Full Deletion/ Duplication

This hereditary detects gross deletions and duplications mutations in BRCA1 and BRCA2 from DNA isolated from a blood specimen.

Testing Method and Background

This is a Multiplex Ligation-dependent Probe Amplification (MLPA) assay that includes three MLPA probe mixes (MRC-Holland, www.mlpa.com): SALSA MLPA P002, P239 (BRCA1) and P090 (BRCA2/CHECK2). This assay only detects common and rare gross deletions and duplications caused by large genomic rearrangements in BRCA1 and BRCA2. As a result, this assay is predicted to miss approximately 80-90% of known cancer-predisposition mutations in BRCA1 and BRCA2, most of which are caused by point mutations or insertions/deletions. The use of this assay as a stand-alone test without previous NGS sequencing is not recommended as this can lead to misinterpretation of test results. This MLPA assay should only be used in combination with next generation sequencing (NGS) analysis for comprehensive testing of BRCA1 and BRCA2 mutations.

This panel analyzes BReast CAncer genes 1 and 2 (BRCA1 and BRCA2), that code for proteins that help repair DNA damage. Inherited mutations in BRCA1 or BRCA2 are associated with autosomal dominant hereditary breast and ovarian cancer (HBOC) syndrome (OMIM ID: 604370) which is characterized by increased lifetime risk for developing breast, ovarian and other types of cancer.

Highlights of BRCA1 and BRCA2 Full Deletion/ Duplication

Targeted Region

BRCA1, BRCA2: Gross deletion and duplications mutations

- **This MLPA assay detects large deletions and duplications in the BRCA1 and BRCA2 genes are usually missed by standard sequence analysis.**
  It is recommended to use this MLPA assay in combinations with sequence analysis since most defects in BRCA1 and BRCA2 are point mutations and will not be detected by MLPA.

Ordering Information

Get started (non-HFHS): Print a Hereditary Cancer Panels requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "BRCA1/2 Deletion/Duplications Only" (DNA2100019)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Saliva specimen - Oragene self-collection kit
- Extracted DNA - from a CLIA-certified Laboratory

Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-10 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81164, G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCPD

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