MAYO CLINIC LABORATORIES

TEST OBSOLETE

Notification Date: May 13, 2019
Effective Date: June 13, 2019

Hereditary Breast Cancer 6 Gene Panel

Test ID: BRST6

Explanation:
Due to low utilization, this test will become obsolete on June 13, 2019.

Recommended Alternative Test:
Hereditary Breast and Colorectal Cancer Panel

Test ID: BRCRC

Methodology:
Custom Sequence Capture and Targeted Next-Generation Sequencing Followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing and Gene Dosage Analysis by Array Comparative Genomic Hybridization (aCGH) or Multiplex Ligation-Dependent Probe Amplification (MLPA)

Genes Included:
BRCA1, *BRCA2*, *TP53*, *PTEN* (including the promoter), *CDH1*, *STK11*, *MLH1*, *MSH2*, *MSH6*, *PMS2*, and *EPCAM*

Useful For:
- Establishing a hereditary susceptibility to cancer
- Evaluation of families with a history suggestive of a predisposition to both breast and colorectal cancer
- Identification of familial mutation to allow for predictive testing in family members

Reference Values:
An interpretive report will be provided.

Specimen Requirements:

Specimen Type:	Whole blood
Container/Tube:	
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood 2. Send specimen in original tube
Minimum Volume:	1 mL

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Refrigerated	

Cautions:

Clinical Correlations:

- Some individuals who have a hereditary susceptibility to breast, endometrial, or colon cancer may have a mutation that is not identified by this method (eg, promoter mutations, deep intronic mutations). The absence of a mutation, therefore, does not eliminate the possibility of a hereditary susceptibility to breast cancer in the individual or family. For predictive testing, it is important to first document the presence of a gene mutation in an affected family member.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete. We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.

Technical Limitations:

- In some cases, DNA variants of undetermined significance may be identified. Due to the limitations of next generation sequencing, we can detect greater than 93% of insertions and deletions up to 20 bases and 43 bases, respectively. If a diagnosis is still suspected, consider full gene sequencing using traditional Sanger methods. Single or multi-exon deletions as well as whole gene deletions will be detected by array comparative genomic hybridization (aCGH) or multiplex ligation-dependent probe amplification (MLPA). Rare polymorphisms exist that could lead to false-negative or false-positive results.
- If results obtained do not match the clinical findings, additional testing should be considered.

Evaluation Tools:

- Multiple in-silico evaluation tools were used to assist in the interpretation of these results. These tools are updated regularly; therefore, changes to these algorithms may result in different predictions for a given alteration. Additionally, the predictability of these tools for the determination of pathogenicity is currently not validated.
- Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

Reclassification of Variants-Policy:

- All detected alterations are evaluated according to American College of Medical Genetics and Genomics recommendations (1). Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.
- At this time, it is not standard practice for the laboratory to systematically re-review likely deleterious alterations or variants of uncertain significance that are detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

CPT Code:
81162 – BRCA1-BRCA2
81321 – PTEN
81405 – STK11
81405 – TP53
81406 – CDH1
81292 – MLH1
81295 – MSH2
81298 – MSH6
81317 – PMS2
81319 – PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants
81403 – EPCAM
81228 – Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, Bacterial Artificial Chromosome [BAC] or oligo-based comparative genomic hybridization [CGH] microarray analysis)
81479 (if appropriate for government payers)

Day(s) Setup:
Performed weekly, varies

Analytic Time:
3 weeks

Questions
Contact Heather Flynn Gilmer or Melissa Lonzo Green, Laboratory Technologist Resource Coordinators, at 800-533-1710.