Henry Ford Health System
CENTER FOR PRECISION DIAGNOSTICS

BRCA1 & *BRCA2* Full Sequencing and Full Deletions/Duplications (2 genes)

BRCA1	*BRCA2*								

Indication

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. Mutations in *BRCA1* and *BRCA2* are responsible for approximately 20 to 25% of hereditary breast cancers and about 5 to 10% of all breast cancers. In addition, mutations in *BRCA1* and *BRCA2* cause around 15% of ovarian cancers. Specific cancer risks are different between *BRCA1* and *BRCA2*. *BRCA1* pathogenic mutations are associated with increased lifetime risk for breast cancer (40-87%), ovarian cancer (16-54%), male breast cancer (1-2%), prostate cancer (up to 20%), and pancreatic cancer (1-3%). *BRCA2* pathogenic mutations are associated with increased lifetime risk for breast cancer (up to 84%), ovarian cancer (up to 27%), male breast cancer (6%), prostate cancer (up to 20%), pancreatic cancer (2-7%), and melanoma.

Hereditary cancer syndrome is a genetic predisposition to develop certain types of cancers, often at an early age. Hereditary cancer risk assessment is performed to identify patients and families who may be at risk. Clues that a hereditary cancer syndrome may be present include the following:

- Cancer diagnosed at an unusually young age (<50 year of age for breast cancer)
- Multiple BRCA-associated cancers in the same person
- Bilateral or multiple primary breast cancers in the same person
- Family history of ovarian, breast, pancreatic, melanoma, or prostate cancer
- Breast or ovarian cancer and Ashkenazi Jewish ethnicity
- Occurrence of certain types of adult cancer in which the probability of harboring a hereditary cancer syndrome is high: triple negative (ER-/PR-/HER2/neu-) breast cancer; ovarian, tubal or peritoneal cancer; male breast cancer

Testing method

Next Generation Sequencing (NGS) provides coverage of all coding exons and noncoding DNA in exon-flanking regions (on average 50 bp) enriched using hybrid capture Illumina TruSight Cancer Sequencing Panel. Single base pair (point) mutations, small insertions/deletions (1-25 bp), complex insertions and deletions, or larger deletions and duplication (<100 bp) are detected using a combination of clinically validated computational data analysis methods for sequence variant calling, filtering, and annotation. Gross deletions and duplications at each targeted gene and exon are evaluated through comparative depth of coverage analysis of NGS targeted sequencing data using clinically-validated analysis algorithm. All reportable copy number variants are confirmed by independent methodology using gene-specific Multiplex Ligation-dependent Probe Amplification (MLPA) assay.

Turnaround time
5-10 business days

Sample requirements
3 ml peripheral blood in EDTA (lavender) top tube
Specimen stability: Ambient - 72 hours; Refrigerated - 1 week

CPT codes
81162, G0452

References

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Ship Specimens to:

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