Prima Cancer Genetics

MEDGENOME

BRCA Testing for Hereditary Breast & Ovarian Cancers

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

Breast and ovarian cancer in Indian women

Annual incidence of approximately

1.4 lakh

new cases of breast cancers in India.

Cancer incidence Indian women to increase from

110 to 190-200

cases per lakh by 2025.

3 times

more cases of familial breast and ovarian cancers are recorded in India, as compared to Western countries.

Mortality to incidence ratio for breast and ovarian cancers with

1/28

women likely to develop it during her lifetime

BRCA gene test

Inherited BRCA gene mutations are responsible for about 5 percent of breast cancers and about 10 to 15 percent of ovarian cancers [1].

BRCA1 or BRCA2 are also the most prominent among the genes associated with Hereditary Breast and Ovarian Cancer (HBOC) Syndrome.

Includes increased risk of developing

- *Fallopian tube cancer*
- *Peritoneal cancer*
- *Pancreatic cancer*
- *Prostate cancer.*

The BRCA gene impact

Cancer type	Status	Population average risk	Risk with BRCA1 mutation	Risk with BRCA2 mutation
Breast	**Unaffected** Risk of developing cancer by the age of 80	12%	46% - 87%	38% - 84%
	Affected Cancer in other breast	2% (Within 5 years)	21.1% (Within 10 years) 83% (By the age of 70)	10.8% (Within 10 years) 62% (By the age of 70)
Male Breast	**Unaffected**	0.1%	1.2%	Upto 8.9%
Ovarian	**Unaffected** Risk of developing cancer by the age of 80	1% - 2%	39% - 63%	16.5% - 27%

BRCA test by MedGenome

NGS based

Uses Next Generation Sequencing based genomic DNA analysis to identify multiple harmful mutations

Mutations Covered

Covers Single Nucleotide Variations (SNVs), short insertions and deletions (InDels), structural variants, copy number variations

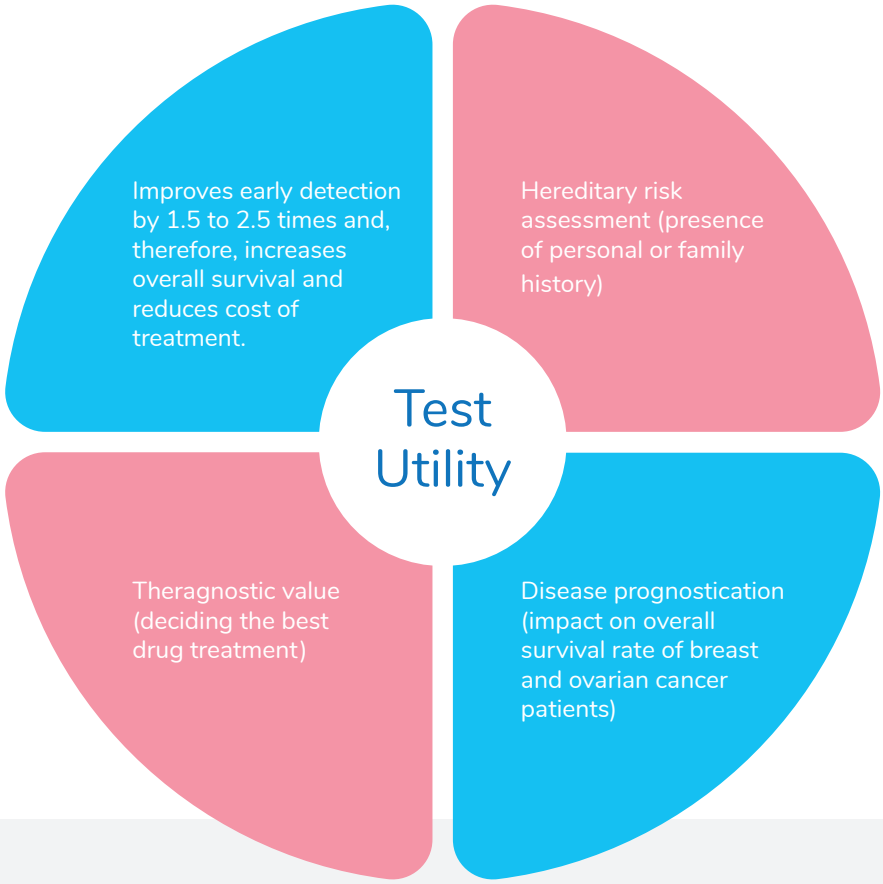
High accuracy and sensitivity

BRCA1 & *BRCA2* genes are covered 100%.
Sensitivity and Specificity to detect SNVs and InDels is 100%

Who should be tested?

Individuals might be at risk of having BRCA gene mutation and can be candidates for BRCA gene testing, if one of the following conditions is fulfilled.

- A personal history of breast or ovarian cancer diagnosed at young age (premenopausal), bilateral breast cancer (affecting both breasts) or the presence of both ovarian and breast cancer
- A family history of breast, ovarian, fallopian tube, peritoneal, prostate, or pancreatic cancer
- A male family member having breast cancer
- A relative with a known deleterious mutation in BRCA1 or BRCA2 genes
- A history of breast cancer diagnosed below age of 45 years
- A family member with bilateral breast cancer below age of 50
- An individual with triple negative breast cancer below age of 60 years with or without family history
- Two or more relatives with ovarian cancer
- Both breast and ovarian cancers in either the same woman or the same family
- Ashkenazi Jewish ethnicity



Test requirements

Sample type	Sample Requirement	Transportation Conditions
Blood	3-5 ml in EDTA tube	20-25 °C
Purified Genomic DNA	1µg high quality DNA (50-100 ng /microlitre)	20-25 °C

Once the sample reaches the laboratory, the specific regions of DNA extracted from blood, will be enriched using an appropriate technology and sequenced in a multiplexed set up. The sequences obtained will be aligned to human reference genome (GRCh37/hg19) using BWA program[4, 5] and analysed using Picard and GATK version 3.6[6, 7] to identify variants relevant to the clinical indication.

Turnaround Time (TAT)

- The time taken for generating a clinical report will be maximum of 3 weeks from receiving the samples in the lab.
- In cases where the sample quality is poor (sample which fails our QC/QA), the TAT will be prolonged. A new batch of the sample will be needed for sequencing and analysis.

References

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3. Kuchenbaecker KB, Hopper JL, Barnes DR, et al. Risks of breast, ovarian, and contralateral breast cancer for BRCA1 and BRCA2 mutation carriers. JAMA 2017; 317(23):2402-2416.
4. Meyer, L.R., et al., The UCSC Genome Browser database: extensions and updates 2013. Nucleic Acids Res, 2013. 41(D1): p. D64-9.
5. McKenna, A., et al., The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. Genome Res, 2010. 20(9): p. 1297-303.
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7. McLaren, W., et al., Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor. Bioinformatics, 2010. 26(16): p. 2069-70.
8. Peshkin BN, DeMarco TA, Brogan BM, Lerman C, Isaacs C. BRCA1/2 testing: Complex themes in result interpretation. Journal of Clinical Oncology 2001; 19(9):2555–2565.

Notes

MEDGENOME

Prima by MedGenome offers a wide range of Oncology and Haematology genetic tests, these include:

Molecular Testing for Hematological Malignancies, Comprehensive Leukemia Panel

Differential Diagnosis

Prognosis

IGHV Gene Mutation Testing for CLL, Comprehensive Leukemia Panel, BCR-ABL1 gene fusion analysis

Hereditary Cancer Panel, BRCA1 and BRCA2 gene test, Thalassemia Mutation Test

Risk Assessment

Therapy Selection

Comprehensive Tumor Gene Panel, Somatic Mutation Panel, Comprehensive Leukemia Panel, Molecular Testing for Lung Cancer

OncoTrack, OncoSelect, OncoFocus (Liquid Biopsy Test)

Surveillance

Therapy Monitoring

NGS based IRMA, BCR- ABL1 gene fusion analysis

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