

NextGen BRCA 1 & 2

Therapeutic Decision Making in Breast and Ovarian Cancer based on BRCA1 and BRCA2 Mutation



BRCA GENE IMPACT

General Population	BRCA 1	BRCA 2	Age	Cancer Type
13%	55%-72%	45%-69%	70-80 years	Breast cancer
1.2%	39%-44%	11%-17%	70-80 years	Ovarian cancer



Breast and Ovarian cancer in Indian Women



1.4 lakh
New cases of
Breast Cancer

Cancer
Incidence
from 100 to
200 per lakh
by 2025



3 Times more
case of familial
Breast and
Ovarian Cancer

Mortality to
Incidence ratio
with 1/28 women
likely to develop
during lifetime





Benefits of BRCA1/2 Genetic testing

- ▶ A positive test result may allow people to make informed decisions about their future health care.



Who Should consider Testing

Family history of



- ▶ Breast, Ovarian, fallopian tube, Peritoneal, Prostate or Pancreatic cancer



- ▶ Breast Cancer diagnosed before age of 45



- ▶ Two or more relatives of Ovarian cancer



- ▶ Individuals with triple negative (Estrogen receptor, progesterone receptor and Her2/neu) breast cancer at < 60 years of age



- ▶ Male Breast cancer



- ▶ Ashkenazi Jewish descent with personal history of Breast, Ovarian or Pancreatic cancer



Variant Categorization

- ▶ Pathogenic
- ▶ Likely Pathogenic
- ▶ VUS (Variants of unknown significance)

Metropolis is the only lab to give conclusive opinion on variants



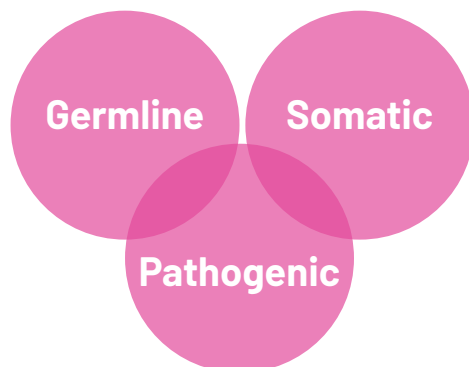
Germline vs Somatic

- ▶ Germline mutations are heritable and present in every cell of a person's body, thus testing only needs to be done once.



Somatic mutations are acquired genetic changes

- Tumor biopsy or surgical specimens, or any other fluid materials that contain malignant cells, e.g., malignant pleural effusion or ascitic fluid



Most Tumor NGS reports are silent on whether an identified pathogenic mutation is germline or somatic in origin¹

- The most conclusive way is to test an accompanying germline sample (e.g., blood)



Tumor BRCA1/2 testing can detect any pathogenic germline mutations

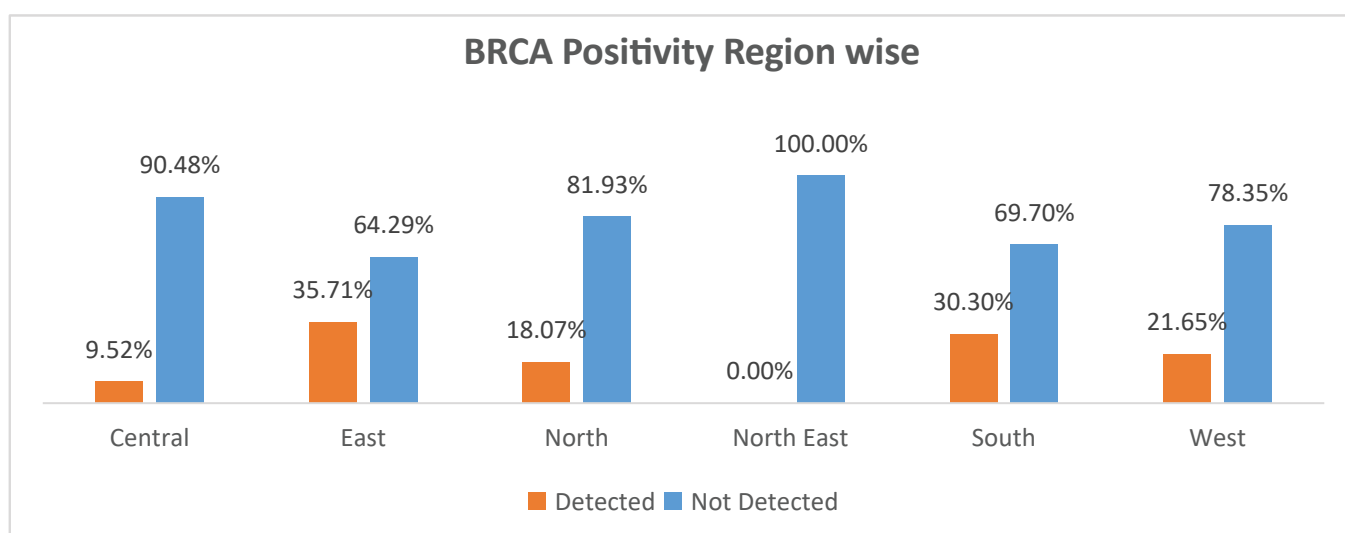
- Low false positive rates
- 90% concordance between somatic and germline variant findings Specimen: pleural effusion, ascitic fluid



Probability of finding an incidental pathogenic BRCA1/2 mutation is ~2–4%, of which ~80% is germline in origin⁴

Metropolis Data

BRCA1 and BRCA2 variants were identified in 151 of 625 individuals (24.16%)





BRCA test by Metropolis



NGS Based: Genomic DNA analysis to identify multiple pathogenic mutations



Mutations Covered: Single Nucleotide variations (SNVs), short insertions deletion (InDels)



High accuracy and sensitivity to detect SNVs and Indels is 100%

Test Utility

Early detection, Genetic predisposition and management of breast cancer

Hereditary risk assessment

Disease Prognostication

Theranostic value (Therapeutics+ Diagnostics)

TEST INFORMATION

Test code	Test/Profile Name	Patient fees in Rs	Test Schedule	Reported on	Method
B0077	NextGen BRCA Comprehensive (NGS & MLPA), Blood	30000	Daily: 7:30 am	4 weeks	NGS & MLPA
B0106	NextGen BRCA Somatic, FFPE tissue	22000	Daily: 7:30 am	25th day	NGS
B1032	NextGen BRCA Germline, Blood	16000	Daily : 9:00am	15th day	NGS

Oncomet Expertise



One stop Solution for Oncology tests



Highly experienced pool of Oncopathologists and Panelist



Capabilities Include Morphology to Molecular



Recipient of 2 Gold Awards for Exceptional contributions to Women's cancer care and Management at cancer care summit and high-end diagnostic services at 8th India health and wellness award

Reference :

1. Wong, Rachel Su Jen, & Soo-Chin Lee. "BRCA sequencing of tumors: understanding its implications in the oncology community." Chinese Clinical Oncology [Online]. 9.5 (2020): 66.
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3. Armstrong N, Ryder S, Forbes C, et al. A systematic review of the international prevalence of BRCA mutation in breast cancer. Clin Epidemiol 2019;11:543-61.
4. Thompson D, Easton DF. Cancer Incidence in BRCA1 Mutation Carriers. J Natl Cancer Inst 2002;94:1358-65.
5. Paul A, Paul S. The breast cancer susceptibility genes (BRCA) in breast and ovarian cancers. Front Biosci (Landmark Ed) 2014;19:605-18.
6. Cavanagh H, Rogers KMA. The role of BRCA1 and BRCA2 mutations in prostate, pancreatic and stomach cancers. Hered Cancer Clin Pract 2015;13:18.

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