Ambry Genetics

RESEARCH FOR YOUR PRACTICE

Casting a Wider Net for Male Breast Cancer Patients

RECENT DATA REVEAL THE NEED TO TEST BEYOND *BRCA1/2*

New research from Ambry Genetics published in Breast Cancer Research and Treatment[1], in collaboration with Mayo Clinic, Huntsman Cancer Institute, and UCI (University of California- Irvine), aims to better describe the genes contributing to inherited risk for male breast cancer (MBC).

WHY THIS MATTERS TO YOU

In the largest reported study of men with breast cancer undergoing multigene panel testing, we offer new insights into genes associated with an increased risk of MBC beyond *BRCA1* and *BRCA2*.

BACKGROUND

Current guidelines recommend genetic testing for *BRCA1* and *BRCA2* in anyone with a personal or family history of MBC[2]. However, genetic predisposition to MBC, particularly beyond *BRCA1/2* is still not well understood.[3] This research aims to further our understanding and explore the utility of multigene panel testing (MGPT) for these patients.

In this study, >700 MBC patients underwent MGPT at Ambry Genetics including at least 16 breast cancer susceptibility genes: *ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MRE11A, NBN, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D*, and *TP53*.

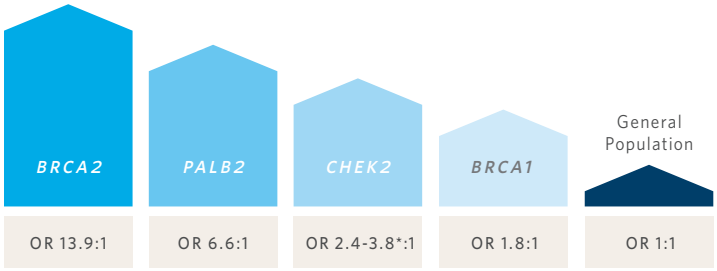
715
Men with breast cancer studied

POINTS FOR YOUR PRACTICE

- In addition to *BRCA1* and *BRCA2* this study suggests *PALB2* and *CHEK2* as a MBC susceptibility gene. Notably, men with *CHEK2* c.1100delC mutations may consider earlier surveillance and/or a higher suspicion for MBC at a younger age
- Men carrying mutations had similar average ages of diagnosis and personal/family histories compared to men without mutations, which supports current NCCN® guidelines recommending genetic testing for patients with MBC at any age, regardless of other personal or family history of cancer
- Identification of germline mutations in MBC patients allows for testing at-risk family members and subsequently adjusting their medical management
- Men with breast cancer should consider MGPT, such as BreastNext, that includes at least *BRCA1, BRCA2, CHEK2* and *PALB2*

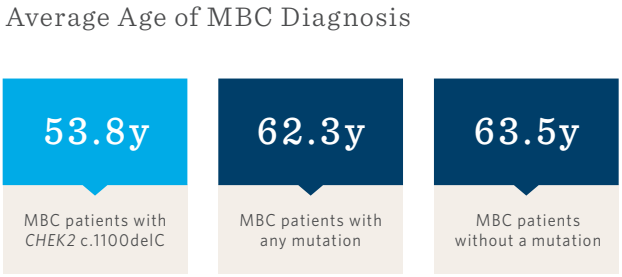
SIGNIFICANT FINDINGS

- 18.1% of men with no prior *BRCA1/2* testing were found to have a breast cancer gene mutation
- *BRCA2* and *CHEK2* mutations were most common
- Odds ratios (OR), or the likelihood of developing male breast cancer, were found to be increased for several genes including *BRCA2*, *CHEK2*, and *PALB2*



**Varies depending on mutations included*
***Confidence intervals (CI) are available for each OR in the publication. See the link "Read the Research" and the citation below.*

- *CHEK2* c.1100delC carriers had a significantly lower age of MBC diagnosis



Learn more about our research here.

REFERENCES

1. Pritzlaff M and Summerour, P, *et al*. Male breast cancer in a multi-gene panel testing cohort: insights and unexpected results. Breast Cancer Res Treat. 2017 Feb;161(3):575-586.
2. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®). Genetic/Familial High-Risk Assessment: Breast and Ovarian. V2.2017.
3. Sousa B, *et al*. An update on male breast cancer and future directions for research and treatment. Eur J Pharmacol. 2013 Oct 5;717(1-3):71-83.