B-GREAT MILESTONES

DECEMBER 2022

Welcome Message

We are excited to share our latest newsletter with you, through which we provide information about the relevance of inherited breast cancer in Black women; and also highlight new research we are conducting that may be of interest. If there are topics you would like for us to cover through these newsletters, please let us know – we can be reached at ICARE@vumc.org. We also want to reiterate our gratitude to many of you who have chosen to participate in our studies – it is because of you that we have been able to contribute to scientific advances, all with the purpose of improving care of those with inherited forms of cancer.

With sincere gratitude,

Sincerely,

Tuya Pal, MD, FACMG *on behalf of the B-GREAT Team*

Want to stay informed about inherited cancers?

Consider enrolling in the Inherited Cancer Registry (ICARE), through which you will:

- Contribute to new discoveries in the field of inherited cancer.
- Get care updates personalized to you as new guidelines are released.
- Find out about other studies for which you may be eligible.

Enroll online now!

Scan the QR code or enroll at https://redcap.link/ICAREconsent

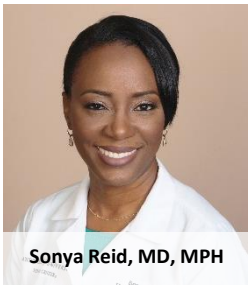
Inherited Breast Cancer in Black Women

How common is it? Prior studies have suggested there may be a higher frequency of mutations in inherited breast cancer genes among Black females with breast cancer in the United States (US), Caribbean, and sub-Saharan Africa.[1-6] In the US, the *BRCA1/2* mutation frequency was reported as 12.4% among young Black females with breast cancer through a Florida cancer registry-based study,[1] similar to a Michigan clinic-based study which reported *BRCA* mutations in 8.1% of Black patients compared to 3.6% of non-Ashkenazi Jewish White patients (p=0.02) with Black patients having mutation prevalence approaching that of Ashkenazi Jewish patients.[2] In studies of unselected breast cancer patients in the Caribbean, *BRCA* mutations were identified in ~23% in the Bahamas[3]; and ~10% in Trinidad and Tobago.[4] In sub-Saharan African studies of unselected breast cancer patients from Nigeria, Uganda, and Cameroon, over 10% had *BRCA* mutations and ~15% had mutations in an inherited breast cancer overall.[5,6] Collectively, these studies suggest the possibility of a higher prevalence of mutations in inherited breast cancer genes among Black populations.

What genes are involved? Studies focused on multi-gene panel testing among women of African ancestry have suggested similar associations of genes with breast cancer risk, as that previously reported among predominantly European ancestry individuals. Specifically, Palmer et al. reported on a case-control study of 10,000 Black females, in whom the frequency of pathogenic variants was 10.3% in those with ER- disease, 5.2% in those with ER+ disease, and 2.3% of controls. Genes associated with the highest risks were *BRCA1* (OR: 47), *BRCA2* (OR: 7.25) and *PALB2* (OR: 8.54).[7] High risk for ER- disease was reported for those with *RAD51D* (OR: 7.82), while moderate risks for ER+ disease were reported in the *CHEK2* and *ATM* genes. Similarly, in a case control study of over 3,000 Black women, significant associations with breast cancer risk were reported for *BRCA1, BRCA2, PALB2, ATM, CHEK2, TP53, NF1, RAD51C,* and *RAD51D*.[8]

[1]Pal et al. Cancer. 2015;121(23):4173-80. PMID: 26287763; [2]Ciuro et al. Clin Breast Cancer. 2021;21(3):e220-e227. PMID: 33168447; [3]Donenberg et al. Breast Cancer Res Treat. 2011;125(2):591-6. PMID: 20838878; [4]Donenberg et al. Breast Cancer Res Treat. 2016;159(1):131-8. PMID: 27469594; [5]Zheng et al. J Clin Oncol. 2018;36(28):2820-2825. PMID: 30130155; [6]Adedokun et al. Cancer Epidemiol Biomarkers Prev. 2020;29(2):359-367. PMID: 31871109; [7]Palmer et al. J Natl Cancer Inst. 2020;112(12):1213-1221 PMID: 32427313; [8]Díaz-Zabala et al. Genet Med. 2022;24(7):1468-1475. PMID: 35396981. Social media post May 17th, 2022. Available at: https://tinyurl.com/post5172022.

Featured Experts in the Field



Sonya Reid, MD, MPH

Breast Cancer Tumor Genomic Studies to Learn More About Breast Cancer Development and Treatment in *BRCA1*, *BRCA2*, *PALB2*, *CHEK2*, and *ATM* Carriers

Together with Dr. Sonya Reid, we are currently conducting a study to learn more about breast cancers that develop in *BRCA1*, *BRCA2*, *PALB2*, *CHEK2*, and *ATM* carriers. This type of study is important, to try to figure out how these tumors develop and how to best treat them. Through this study, we are doing free genomic testing on tumor and blood samples. Please scan the QR code to enroll online or visit https://redcap.link/ICAREconsent.

Mya Roberson, PhD

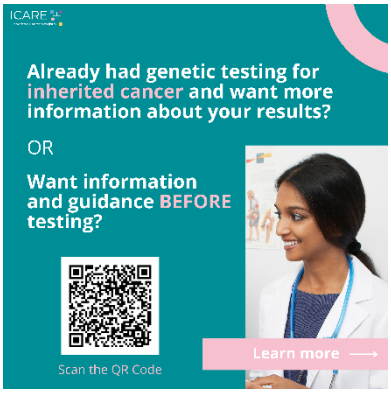
Interested in Getting Genetic Counseling?

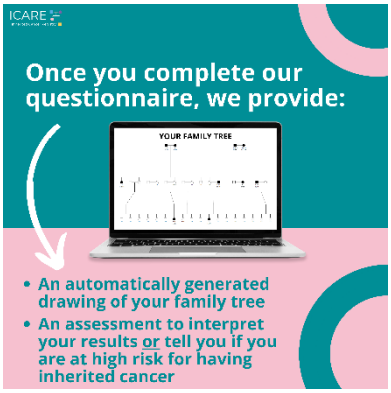
A recent paper published in *JAMA Health Forum* by Dr. Mya Roberson showed that many large private health insurers cover the cost of genetic counseling. Learn more at: https://tinyurl.com/RobersonArticle.

Dr. Roberson is expanding her efforts to better understand the disparities that exist for genetic testing in both the tumor and blood, to guide strategies to figure out ways that testing can become accessible across all populations.

Tool for Inherited Cancer Predisposition Counseling and Testing (TIPS)

Enroll today to receive free education and assessment for inherited cancer risk!

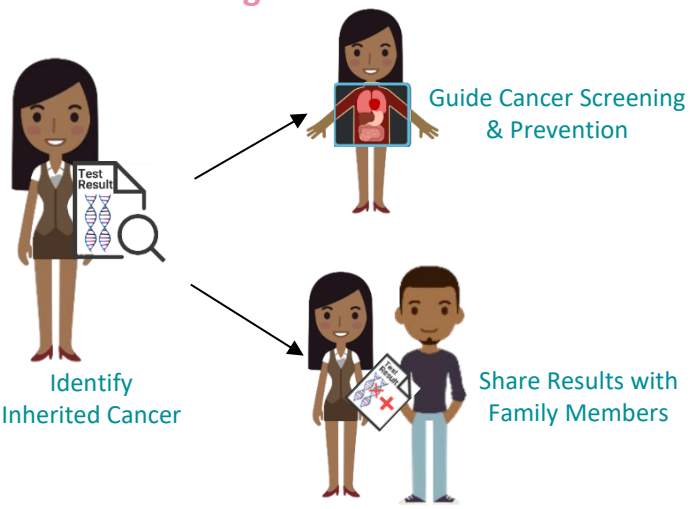




IMProving Care After Inherited Cancer Testing (IMPACT) Study

Do you have a mutation (positive result) in an inherited cancer gene? Now what?

It may be hard to keep up with the many medical appointments and recommendations to manage higher risks of cancer, especially when guidelines keep changing as new research becomes available. As well, even though we know this information may also be helpful for family members to know about, it is sometimes hard to share these results with family, explain why this information is important to family members, and how they can get tested if they want. To address this need, we are actively recruiting to a clinical trial (called our "IMPACT Study") to test some ways to help patients stay updated about how to manage cancer risks and share information with family members. Please consider participating in our Inherited Cancer Registry (ICARE) through which we are actively recruiting to the IMPACT Study.



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