BASSER CENTER FOR BRCA

Penn Medicine

Disparities in Genetic Testing for Cancer Susceptibility

By:

Susan M. Domchek, MD
Executive Director, Basser Center for BRCA at Penn Medicine's Abramson Cancer Center

This year marks the 25th anniversary of the cloning of the *BRCA1* gene. This incredible milestone allowed us to understand the functions of *BRCA1* and *BRCA2* along with the cancer risks associated with mutations in these genes. Today, innovations in research have illuminated the path to lifesaving options for individuals and families affected by *BRCA* mutations, such as improved cancer screenings, risk reducing surgeries, and targeted treatments. We've made so much progress, yet many carriers do not know their status and therefore cannot take advantage of the opportunity to reduce their cancer risk or detect it early. Underutilization of genetic testing appears to have many different causes. These span from lack of awareness of the importance of one's family history, lack of discussion with one's healthcare provider, a misunderstanding of the costs of genetic counseling and/or testing, confusion about the complicated field of genetics, and challenges navigating the health care system.

Although individuals who are of Ashkenazi Jewish descent are 10 times more likely to have a mutation (1 in 40), *BRCA1* and *BRCA2* mutations are found in all races and ethnicities. Several studies have shown that even in populations at the highest risk of having *BRCA1/2* mutations (for instance, ovarian cancer patients) many individuals do not undergo testing. Barriers to uptake of genetic testing seem to be a particular issue in Black and Latino communities. Individuals in these communities are at particular risk for missing opportunities for early detection and prevention.

During breast cancer awareness month in October, Mathew Knowles, Beyoncé's father, spoke out about his *BRCA*-related breast cancer diagnosis. His story is incredibly impactful and

important in three key ways: Mr. Knowles is telling us 1) men can get breast cancer, 2) men can inherit *BRCA* mutations and these mutations can be passed on to one's children, and 3) health disparities in the Black community cannot be tolerated. Black women, who are more frequently diagnosed with triple negative breast cancer than white women, are vastly under-tested for genetic mutations. A study at the Ohio State University's James Comprehensive Cancer Center found that only 20% of Black women had undergone testing compared to 67% White women (Padamsee, Meadows and Hills, 2018). The authors conclude, "These findings suggest that information gathering is more complex than has previously been addressed, that information access and provider access are closely related, and that African American women may be systematically disadvantaged with respect to information-generating experiences." Patient awareness is undoubtedly a major factor in testing disparities, as noted in a recent study reporting that over half of White women had awareness of *BRCA* testing compared to 11.4% of Black women and 30% of women of other racial/ethnic groups (Rubinsak et al., 2019). In this study, the authors conclude, "Interest in genetic testing among women in the general population is high. Despite interest, awareness of *BRCA* is poor among Black and Hispanic women even when adjusting for education level."

Within the Latino population, awareness of hereditary cancer risks and genetic testing is low, despite evidence that Latinas have the second-highest prevalence of *BRCA1/2* mutations after Ashkenazi Jews (Weitzel et al., 2013). Reports repeatedly show that Black and Latina women are more likely to be diagnosed with breast cancer at an advanced stage, and they are more likely to die from breast cancer than non-Hispanic White women (American Cancer Society, 2015; Richardson et al., 2016). It is imperative, therefore, that we strive to reduce barriers related to awareness, health literacy, and cultural beliefs and preferences in order to help people from all races and ethnicities accurately assess their risk.

The Basser Center for BRCA is seeking to do just that. We've recently partnered with Alejandra Campoverdi, former aide in the Obama Administration, a *BRCA2* carrier and Latina, to help patients within the Latino community learn about and navigate the very complex world that is cancer genetics. The Basser Center for BRCA's LATINX & BRCA initiative offers educational resources in Spanish, assistance with finding a Spanish-speaking genetic counselor, and ultimately, a community of support. The Basser Center has also started a poster campaign that includes a number of patient education materials in English and Spanish, to spread awareness across the country. As part of this initiative, the Basser Center has collaborated with organizations like the National Society for Genetic Counselors and the ÁRBOLES Familiares Program to expand these important efforts by engaging healthcare providers working with Latino patients and families.

Other projects are underway at the Basser Center including focused outreach to engage the Black community in learning about their hereditary cancer risk. We believe that patients and families armed with education and resources tailored to their unique needs and values will be best equipped to take prompt action to reduce their risk.

Today more than ever, there is tremendous hope for those affected by a BRCA mutation. Thanks to over 25 years of breakthrough research, innovation and collaboration, we now have greater understanding about the steps people can take to detect cancers early or prevent them altogether. However, with as many as 90% of *BRCA1/2* not knowing their status, it is crucial that we continue to help family and community members understand their family history and individual risks through education and support. With the help of organizations such as Oneinforty, more individuals will know their risk.

To learn more about the Basser Center for BRCA, its outreach initiatives, and educational resources please visit us at basser.org, basser.org/latinxbrca and basser.org/blackbrca.

References:

Padamsee TJ, Meadows R, Hills M. Layers of information: interacting constraints on breast cancer risk-management by high-risk African American women, *Ethnicity & Health*. 2018. DOI: 10.1080/13557858.2018.1562053

American Cancer Society. Cancer facts and figures for Hispanics/Latinos 2015-2017. Atlanta: American Cancer Society. 2015; https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/cancer-facts-and-figures-for-hispanics-and-latinos/cancer-facts-and-figures-for-hispanics-and-latinos-2015-2017.pdf

Richardson LC, Henley SJ, Miller JW, Massetti G, Thomas CC. Patterns and trends in age-specific Black-White differences in breast cancer incidence and mortality – United States, 1999–2014. *Morbidity and Mortality Weekly Report.* 2016; 65:1093–1098. DOI: http://dx.doi.org/10.15585/mmwr.mm6540a1

Rubinsak LA, Kleinman A, Quillin, J, Gordon SW, Sullivan SA, Sutton AL, Sheppard V, Temkin SM (2019): Awareness and acceptability of population-based screening for pathogenic BRCA variants: Do race and ethnicity matter? *Gynecologic oncology.* 2019; 154(2): 383-387. DOI: https://doi.org/10.1016/j.ygyno.2019.06.009

Weitzel JN, Clague J, Martir-Negron A, Ogaz R, Herzog J, Ricker C, Jungbluth C, Cina C, Duncan P, Unzeitig G, Saldivar JS, Beattie M, Feldman N, Sand S, Port D, Barragan DI, John EM, Neuhausen SL, Larson GP. Prevalence and type of BRCA mutations in Hispanics undergoing genetic cancer risk assessment in the southwestern United States: a report from the Clinical Cancer Genetics Community Research Network. *Journal of Clinical Oncology*. 2013; 31(31): 1702. DOI: 10.1200/JCO.2011.41.0027