

Disparities in Cancer Genetic Risk Assessment and Testing

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Scientific and technologic advances in genomics have revolutionized genetic counseling and testing, targeted therapy, and cancer screening and prevention (Weitzel, Blazer, MacDonald, Culver, & Offit, 2011). Evidence-based practice guidelines for genetic risk assessment and testing are well established (Scalia-Wilbur, Colins, Penson, & Dizon, 2016). The most commonly referenced hereditary cancer syndrome is hereditary breast and ovarian cancer (HBOC) syndrome caused predominately by gene mutations in *BRCA1* or *BRCA2*. Other high- or moderate-risk genes also associated with HBOC include mutations in the *ATM*, *CDH1*, *CHEK2*, *PALB2*, *PTEN*, *STK11*, and *TP53* genes (National Comprehensive Cancer Network [NCCN], 2016). The identification of a pathogenic mutation in *BRCA1/2* infers an increased risk for a host of cancers for men and women in addition to breast and ovarian cancers; these include melanoma, as well as prostate and pancreatic cancers (NCCN, 2016). Genetic testing results can be the catalyst for patients to access targeted diagnostic (Smith et al., 2015), prevention (Domchek et al., 2010), and treatment strategies (Balmaña, Domchek, Tutt, & Garber, 2011) not routinely recommended to the general population. Among younger women, African American and Hispanic women have a higher rate of cancers that are associated with hereditary

cancer risk, such as triple-negative breast cancer, which is linked to poorer outcomes (Reynolds, 2007). Therefore, genetic testing is particularly important in diverse populations. Unfortunately, all races and ethnic groups are not well represented in current genetic testing practices, leading to disparities in cancer prevention and early detection.

Racial and Ethnic Disparities

Although the awareness (Mai et al., 2014) and use (Rosenberg et al., 2016) of genetic testing in specific populations have increased over time, racial and socioeconomic disparities in access to HBOC risk assessment, counseling, and genetic testing continue to exist in the United States (Daly & Olopade, 2015). In a large national health services study focusing on *BRCA1/2* genetic testing, only 12% of African American and 18% of Hispanic individuals had genetic testing for *BRCA1/2*, compared to 34% of non-Jewish Caucasian individuals (Levy et al., 2011). These disparities have been established for more than a decade (Armstrong, Micco, Carney, Stopfer, & Putt, 2005; Hall & Olopade, 2006; Levy et al., 2011) and persist today (Mai et al., 2014; Yusuf et al., 2015). The lack of genetic counseling and testing in disparate populations has a detrimental cascade effect. Insufficient risk assessment and genetic testing may