

Disparities in Cancer Genetic Risk Assessment and Testing

Meghan L. Underhill, PhD, RN, AOCNS®, Tarsha Jones, PhD, RN, APHN-BC, and Karleen Habin, MSN, RN, CNS

Underhill is an instructor and Jones is a postdoctoral research fellow, both at the Dana-Farber Cancer Institute, and Habin is a research nursing administrator at Massachusetts General Hospital, all in Boston, MA.

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Underhill can be reached at meghan_underhill@dfci.harvard.edu, with copy to editor at ONFEditor@ons.org.

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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

TABLE 1. Selected Guidelines Supporting Cancer Genetic Risk Assessment

Organization	Description	Website
American Society of Clinical Oncology	A specific genetics toolkit is available, in addition to policy statements, as a resource for implementing cancer risk assessment into practice.	http://bit.ly/24wGE30
National Comprehensive Cancer Network	Specific guidelines are provided for hereditary breast, ovarian, and colon cancer risk assessment. Embedded within the treatment guidelines is information for tumor-specific genetic testing.	http://bit.ly/1TXEydL
National Society of Genetic Counselors	Specific practice guidelines outline when to refer individuals for genetic risk assessment and testing.	http://bit.ly/22MMAqn
U.S. Preventive Services Task Force	Recommendations for genetic risk assessment and testing in women at high risk for hereditary breast or ovarian cancer are offered.	http://bit.ly/1Lfn1bb

lead to poor awareness of risk and, subsequently, inappropriate or inadequate early detection, prevention, and treatment services, resulting in poor patient outcomes (Hall & Olopade, 2006).

The low use of genetic testing in underrepresented minority participants compared to Caucasians reduces the generalizability of genetic discoveries and also leads to challenges in interpreting genetic testing results. For example, one study of 1,765 women found that African Americans are more likely to have a *BRCA1/2* genetic testing result of a variant of uncertain significance (VUS) (45%), in which a genetic change is found but clinical significance is not understood, compared to Caucasian participants with a VUS rate of about 6% (Opatt, Morrow, & Daly, 2006). Because fewer racially and ethnically diverse individuals have taken part in genetic testing, genetic diversity is not represented in the data available to interpret genetic testing results, creating challenges in the efficacy of genetic testing and clinical management (Saulsberry & Terry, 2013). This disparity in testing ultimately may advance the science for some but not all populations, widening the health disparities gap (Cohn, Husamudeen, Larson, & Williams, 2015).

Factors Associated With Genetic Risk Assessment and Testing

Patient-level factors: At the patient level, demographic, clinical, and psychosocial factors are associated with awareness and use of cancer genetic risk assessment and testing. Despite hereditary risk affecting both genders, in all racial groups, women are more likely to be aware of cancer genetic testing than men (Mai et al., 2014). Age has been found to affect awareness of cancer genetic testing; individuals aged 25–39 years or individuals aged older than 60 years are less likely to be aware of genetic testing (Mai et al., 2014). In addition, having a personal cancer diagnosis or a diagnosis of triple-negative breast cancer is associated with the use of genetic testing in young African American women (Cragun et al., 2015). Having a family history of cancer diagnoses is also associated with increased awareness of genetic testing (Mai et al., 2014).

Although personal demographic factors are non-modifiable, some factors are variable and associated with awareness or use of cancer genetic risk assessment and testing. Having less education, or being educated at the high school level or lower, is associated with reduced awareness (Mai et al., 2014) and use (Cragun et al., 2015; Olaya et al.,

2009) of cancer risk assessment and testing. In addition, medical mistrust is associated with less use of cancer risk assessment and genetic testing (Sheppard, Mays, LaVeist, & Tercyak, 2013). Individuals with more self-efficacy or confidence are more likely to engage in these services (Sheppard et al., 2013).

Provider-level factors: Being referred for genetic risk assessment or testing by a healthcare provider is a factor associated with genetic testing use in young African American women with breast cancer (Cragun et al., 2015). However, a dearth of available cancer genetic services exists in some parts of the country and is related to the limited number of healthcare providers adequately trained in the emerging field of genetics (Weitzel et al., 2011). Scant knowledge among physicians about who should be referred, the value of referral, and how to refer also contribute to low referral levels (Weitzel et al., 2011). Although minority populations are interested in learning about genetic testing, they are less likely to report receiving a referral for genetic services and are more likely to report unmet needs (Jagsi et al., 2015).

Health system-level factors: When genetic risk assessment and testing are available, lack of or insufficient health insurance coverage for genetic consultations, genetic

testing, and recommended follow-up care are major barriers (Weitzel et al., 2011). Individuals with private insurance (versus public insurance) are more likely to get genetic testing (Cragun et al., 2015). Even with insurance, minorities may be less likely to have genetic testing. In one study of 213 highly insured individuals who underwent genetic testing, 63% were Caucasian, 15% were Hispanic, 7% were African American, and 5% were Asian (Olaya et al., 2009). Costs associated with cancer risk assessment and genetic testing were found to be important to Latina women, determining if they would undergo genetic testing with high costs being a barrier (Gammon et al., 2011).

Implications for Nursing

Genetic testing is an important component of comprehensive oncology care at all levels, from early detection to treatment. Nurses in all settings have a pivotal role to play in supporting patients who would benefit from cancer genetic

risk assessment or testing. Being informed and aware of risk factors for hereditary cancer syndromes is an important competency of all practicing nurses (Jenkins et al., 2015). Nurses should recognize and be aware of red flags for hereditary cancer risk and risk factors associated with cancer (NCCN, 2016) by referring to established clinical guidelines. Just as genetic testing for HBOC is expanding beyond *BRCA1/2*, the importance of cancer syndromes other than HBOC is well established (Hall & Olopade, 2006; Karlitz et al., 2015). Incorporating risk assessment for all cancer syndromes, such as hereditary colon cancer risk (NCCN, 2015), is an important part of the family history assessment process. Table 1 lists clinical resources for nurses looking to incorporate hereditary cancer risk assessment into their practice.

Helping patients to identify their risk for hereditary cancer, beginning with an in-depth three-generation family history and personal health history, is a key step

in improving access to risk assessment and genetic testing (Estrada, LeGrazie, McKamie, & Montgomery, 2015). Without an accurate assessment of personal and family cancer risk, individuals in need of cancer risk assessment and genetic testing may not be appropriately referred. Nurses are at the frontline of patient assessment and are obligated to ensure that accurate and current family health histories are obtained for all patients. Once risk is identified, nurses can help to ensure that patients and families are able to access services to assist them in obtaining appropriate genetics care. Great potential exists for oncology nurse navigators in genetics to help those who are underserved access much-needed care and follow-up (McAllister & Schmitt, 2015).

Table 2 provides some publically available financial and supportive care resources for patients. Nurses caring for patients in need of genetic services should contact the local department of public health or local breast cancer advocacy agencies for available local- and

TABLE 2. Patient Resources for Genetic Information, Risk Assessment, and Testing

Organization	Description	Contact Information
Bright Pink	The PinkPal program matches women aged 18–45 years at increased or high risk for breast and ovarian cancer with a peer with similar risk and experiences. With the Ask a Genetic Counselor resource, patients can submit questions, and a certified counselor from InformedDNA will address them.	http://bit.ly/247DSSB (PinkPal program web page); http://bit.ly/1f2WmRL (Ask a Genetic Counselor web page); 312-787-4412 (phone)
Facing Our Risk of Cancer Empowered	For patients and providers with information focused on hereditary breast and ovarian cancer risk	www.facingourrisk.org (website); 866-288-7475 (toll-free helpline); info@facingourrisk.org (email)
Lynch Syndrome International	Provides support and information for patients, families, and healthcare providers dealing with Lynch syndrome	www.lynchcancers.com (website); 203-779-5034 (phone)
Know:BRCA	Intended for patients to learn about their risk for having a <i>BRCA</i> mutation	www.knowbrca.org (website)
Sisters Network Inc.	Provides services to African American women facing financial challenges	http://bit.ly/1s9ZGkG (website); 866-781-1808 (phone); infonet@sistersnetworkinc.org (email)
Susan G. Komen for the Cure	Offers various assistance and support services, including a breast care helpline	http://sgk.mn/1sPXBuM ("Understanding Assistance and Support" web page); 877-465-6636 (helpline)

Note. Contact local and state public health departments for additional resources.

state-level resources. In addition, individual clinics, hospitals, or health systems may have funding from private resources for genetic risk assessment and testing services for uninsured or underinsured persons; they may also be involved in research aimed at improving disparities in the use of genetic services. Oncology nurses need to familiarize themselves with local resources and collaborate with credentialed genetics professionals to identify financial resources to help patients access genetic testing.

In addition to advocating for patients at the clinical level, nurses have a role in health policy coverage of genetic testing. Nurses at all levels can advocate for evidence-based payer coverage of genetic testing at the public level, which often helps address the cost barrier to testing for racially and ethnically diverse populations (Prince, 2015).

Genomics and cancer genetics have rapidly evolved beyond *BRCA1/2* testing alone. Multigene panel testing is now a routine part of clinical genetic testing. Multigene panel testing analyzes genes in addition to *BRCA1/2* that are associated with breast and ovarian cancer risk, as well as risk for other cancers (Desmond et al., 2015). The rate of uncertain results in multigene panel tests can be significant—even as high as 42% (Frey et al., 2015)—further confounding the already high VUS rates in racially and ethnically diverse groups, making intervention and management difficult. Therefore, now more than ever, nurses must engage high-risk and racially or ethnically diverse individuals in genetic services or research to help inform the expanding science of genetic risk. One means to better understand the clinical significance of these variants is to enroll patients in a registry to pool data across the population. The Prospective Registry of MultiPlex Testing (www.promptinfo.squarespace

.com) is a research registry aimed at gathering data to ultimately reclassify variants. Patients who enroll also receive updates about new findings. Oncology nurses can discuss this registry with patients and encourage them to enroll in the registry to help advance information about genetic variants, particularly in minority populations. Patients and families need continual education and updates about the meaning of a VUS. They also have the opportunity to enroll in reputable registries and variant classification studies to not only obtain data that may be helpful for their family, but also to move the science of variant reclassification to better represent minority populations.

Conclusion

Cancer genetics risk assessment and testing provides an opportunity to identify at-risk individuals in need of advanced diagnostic, preventative, and curative care. Disparities in awareness and access to genetic testing exist because of multifactorial reasons, of which many are modifiable. Nurses in all practice settings and at all levels have an obligation to ensure that family history is assessed and that patients from all backgrounds are advocated for in the evolving landscape of cancer genetic risk assessment and testing.

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Authorship Opportunity

Genetics & Genomics aims to educate oncology nurses about the emerging role of genetics and genomics in cancer care. Possible submissions include, but are not limited to, application of genetics and genomics in clinical practice, screening and surveillance, case studies to present new ideas or challenge current notions, and ethical issues. Manuscripts should be 1,000–1,500 words, exclusive of tables and figures, and accompanied by a cover letter requesting consideration for this feature. For more information, contact Associate Editor Suzanne M. Mahon, RN, DNSc, AOCN®, APNG, at mahonsm@slu.edu.