Genetic Testing

How genetics and genomics can affect healthcare disparities

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Technological advancements in oncology have influenced improvements in survivorship care to such an extent that healthcare providers’ views have been transformed to align with chronic illnesses. These improvements have been attributed to advances that include increased sensitivity and specificity of diagnostics, delivery of personalized medicine, and specialized oncology training (Asare, Flannery, & Kamen, 2017). The positive effects of cancer programs and advances in care have been observed by greater survivorship rates across cancer diagnoses (Siegel, Miller, & Jemal, 2017). To further illustrate, 20% of new primary cancers occur in survivors of previous primary cancers (Donin et al., 2016).

The Oncology Nursing Society’s ([ONS’s], 2017) position statement on access to quality care calls for nurses, inter-disciplinary healthcare teams, scientists, and policymakers to provide patient-centered cancer care to all people “without discrimination, including populations who are at risk, vulnerable, underserved or underrepresented” (para. 3). Although improvements in survivorship have been observed, disparities in survivorship exist (Seigel et al., 2017). Issues with access to health care (uninsured or underinsured and limited healthcare availability) have long been cited as primary causes for disparities observed in those at-risk populations (Cheek & Howington, 2017). At-risk and vulnerable populations are less likely to use screening services (Asare et al., 2017), participate in cancer prevention programs (ONS, 2017), and be diagnosed at later stages, which can be more difficult to treat (Long, Liu, & Bristow, 2013). They also may be offered inadequate treatment approaches (Torgeson, Boothe, Poppe, Suneja, & Gaffney, 2017) because of age (Sagne et al., 2014) or socioeconomic factors (Long et al., 2013). In addition, many diverse cultural, racial, and ethnic populations also hesitate to undergo genetics and genomics testing (Underhill, Jones, & Habin, 2016).

However, research using big data sets for genome-wide association studies have contributed significant knowledge during the past few years. These combined data sets have permitted analyses with confidence in results that otherwise may not have been elucidated or substantiated in smaller sample sizes (Gao, Pierce, Olopade, Im, & Huo, 2017). As a result, genomic developments have shown the association of specific mutations and cancer survivorship based on race/ethnicity. This article will discuss mutation findings using TP53 as an exemplar, implications for practice, and the role oncology nurses may play in educating the public as a means to reduce healthcare disparities.

TP53 Mutations

Located on the 17th chromosome, tumor protein p53 (TP53) is a tumor suppressor gene inhibiting the development of cancer. The p53 protein consists of four domains, with responsibilities that include transcription factor activation, recognition of DNA sequences, and recognition of DNA damage. p53 plays a major role in apoptosis, growth arrest, DNA repair, and...
inhibition of angiogenesis. Since its initial identification, it has been identified as the most frequently mutated gene (greater than 50%) and implicated in the proliferation of many cancers, including breast, prostate, endometrial, pancreatic, and colorectal (Surget, Khoury, & Bourdon, 2014).

Gao et al. (2017) used gene-level, expression-based analytics to examine the predictive breast cancer risk for estrogen receptor-negative breast cancer. They found that five single nucleotide polymorphisms (SNPs) involving TP53INP2 were significantly associated with the risk of developing estrogen receptor-negative breast cancer. This genome-wide association study used four databases of European, Asian, and African ancestry to look beyond racial disparities for predictive risk and then confirmed the analysis with a smaller database.

Using data from the Surveillance, Epidemiology, and End Results program and the Cancer Genome Atlas to explore racial disparities in patients presenting with endometrial cancer, Tarney et al. (2017) found that Black women presented at an earlier age with more aggressive cancers, worse prognostic factors, and worse survival. The sample represented a total of 86,702 women; 90.2% were White, and only 9.8% were Black. Survival was worse across all ages for Black women diagnosed with endometrial cancers than for White women. At the time of diagnosis for women older than age 50 years, Black women had a greater than 50% chance of having a high-risk histology, and White women had a 25% risk for high-risk histology. TP53 mutations were more common in Black women than White women and were associated with worse outcomes; however, these mutations were observed to present with increasing age, and the associated racial disparity became equivocal.

Weige et al. (2014) explored the association of differences in variant allele frequency (proline or arginine) of TP53 with racial disparities and cancer risk. Proline variants have been observed to enhance inflammation, and arginine induces apoptosis. Those researchers found that the TP53Pro72Arg polymorphism of proline/proline was associated with aggressive colon cancer proliferation at younger ages in African Americans. Using a smaller database, Wu et al. (2016) reported similar racial disparities. They found significantly higher rates of TP53 mutations in Hispanic and African American patients. In addition, TP53 mutations were significantly associated with head and neck cancer mortality in the Hispanic cohort. Finally, Murphy et al. (2017) advanced their mouse model of a TP53 mutation, TP53Pro47Ser, that demonstrated the serine variant had significant impairment to its ability to induce programmed cell death and increased cancer risk; this variant was found to increase risk in younger African American women (premenopausal ages).

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Synthesis of these studies' results suggest that TP53 mutations occur with greater frequency in non-White populations and are associated with more aggressive cancers that have poorer prognosis, may be more difficult to treat, and occur at an earlier age. Because the studies reported within this article represent a very small number of those performed exploring genetic associations or predispositions to cancer, it should be noted that racial and ethnic disparities have been observed in additional populations than those addressed here.

**Role of Oncology Nurses**

Cancer statistics (Siegel et al., 2017) indicate a 25% decline in overall cancer deaths from 1991–2014. Although cancer deaths remained higher in Black populations (15% higher than cancer deaths in White populations), the greatest disparity improvement since 2010 has been the reduction in uninsured patients. From 2010–2015, uninsured patients diagnosed with cancer declined from 21% to 11% for Black patients and from 31% to 16% for Hispanic patients.

The exemplars presented in this article demonstrate the value of genetics/genomics-based approaches to examine cancer risks or specific features (e.g., aggressiveness). When used appropriately, healthcare providers can explore an individual's potential risk for developing cancer with more certainty and offer appropriate education regarding preventive strategies and screening opportunities. For those with a new diagnosis of cancer, application of genetic or genomic de-
treatments. Through advances in genetics/genomics, healthcare providers have opportunities to further reduce cancer-related deaths, particularly in diverse populations.

Using the social determinants of health framework (Asare et al., 2017), nurses and diverse communities can be empowered to perform activities aimed to reduce cancer disparities. First, oncology nurses should keep up with advances in genetics and genomics and their impact on care to educate the public, particularly at-risk populations. Second, oncology nurses should work with at-risk, vulnerable, underrepresented, and underinsured populations within their communities to hear concerns about screening and care and to provide a voice through advocacy. Third, oncology nurses should provide preventive strategies that disadvantaged communities can implement within the purview of their cultural priorities. This includes connecting community lay leaders and oncology nurse navigators to perform health histories that offer cancer risk assessments and appropriate genetic testing for those at risk; these connections facilitate incorporation of community values and meaning into public education aimed for that community. Lastly, oncology nurses should advocate for appropriate treatment strategies, including personalized medicine, to be offered so that all individuals receive the best treatment option to improve cancer survivorship (ONS, 2017).

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