Genetic Testing

How genetics and genomics can affect healthcare disparities

Deborah “Hutch” Allen, PhD, RN, CNS, FNP-BC, AOCNP*

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, interdisciplinary 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, Sunjea, & 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...