Pharmacogenomic Germline Testing: Applications in Oncology Nursing

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Since the completion of the Human Genome Project in 2003, genomic research has rapidly advanced. Technological advances and plummeting testing costs have allowed genetic testing to be implemented into daily clinical practice and have deepened the understanding of how genes affect health (Cecchin & Stocco, 2020; Prokop et al., 2018). One current area of genetic applications in clinical practice is precision medicine. Also known as personalized medicine, precision medicine considers an individual’s genetic composition, environment, and lifestyle choices when making decisions regarding disease prevention and treatment (Dodson, 2017).

Pharmacogenetics and pharmacogenomics have become important components of precision medicine. These terms are often used interchangeably. However, pharmacogenetics is generally defined as the study of inherited gene variants that can affect how a person responds to drugs, whereas pharmacogenomics is a broader term describing the study of acquired and inherited variants across the entire genome (an individual’s complete set of genes) and their effects on drug metabolism (Dodson, 2017).

Pharmacogenomics studies how changes to the genome alter proteins involved in drug metabolism, thereby affecting the pharmacokinetics of drugs (Crisafulli et al., 2019). Oncology nurses can be leaders in implementing pharmacogenomics into daily clinical practice. To do so effectively, oncology nurses must understand how DNA is collected and tested, how to interpret the test results, and how to educate patients about their results.

Pharmacogenomic Specimen Collection, Testing, and Results

Pharmacogenomic testing can be performed on normal cells in the body (known as germline testing) and on tumor cells (known as somatic testing). Next-generation sequencing of cancer cells has become commonplace and has facilitated the creation of a growing number of therapies targeting specific genetic variants. This article focuses on germline testing as it relates to genes involved in drug metabolism. In the clinical setting, pharmacogenomic germline testing is used to gain information on how a patient metabolizes specific medications.

Specimen Collection

Three common methods used to collect DNA samples from a patient are through obtaining blood samples, saliva samples, or buccal swabs.