Response to "Women's Knowledge of Genomic Testing and Precision Medicine in Breast Cancer Treatment Decision-Making"

We would like to thank authors Robles-Rodriguez et al. (2024) for their recent article on patient perceptions and knowledge of genomic science in the treatment of breast cancer. The authors appropriately note that physicians may lack knowledge about genomic science, as it is not covered extensively in medical training. The same is true of nursing education (Thomas et al., 2023). Additionally, ever-expanding applications of genomics in cancer care have led to improved survival and expanded treatment options; however, the knowledge and competence of the cancer care team to apply the science, order and interpret correct tests, analyze test results, and share information with patients and families for shared decision-making is complex and daunting and creates confusion for patients and families. Correct and consistent use of terminology in this complex area is essential.

We would like to note that some of the genomic terms may not have been accurately defined. Robles-Rodriguez et al. (2024) state: "Genetic testing checks for variants or changes in a person's DNA, but genomic testing examines a tumor's molecular composition. Advances in genomic testing have led the way toward precision medicine, a treatment that is specific to a tumor's genetic polymorphism" (p. 199).

The authors are describing germline and somatic (tumor) testing. Germline testing identifies variants associated with inherited cancer risk, and somatic testing identifies acquired variants in the tumor. The difference between genetics and genomics is in the amount of material analyzed. Genetics typically looks at one or a few genes, and genomics looks at the entire genome or all of an individual's DNA and interaction with environmental factors. It is possible to analyze the entire human germline genome to better understand inherited risk for developing diseases.

A polymorphism is a variant with a frequency in the general population of greater than 1% (Richards et al., 2015). The use of the term *polymorphism* is not recommended, as it does not clarify the clinical meaning of a variant.

KEYWORDS cancer; genomics; genomic science; breast cancer; cancer care; germline testing; somatic testing

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Germline variants are classified by pathogenicity (benign, likely benign, unknown significance, likely pathogenic, and pathogenic). Somatic variants are classified in tiers by actionability. Tier I lists variants with strong clinical significance in diagnosis, prognosis, or treatment and are included in clinical practice guidelines. Variants of potential clinical significance are classified as tier II, variants of unknown significance as tier III, and benign or likely benign as tier IV.

Precision medicine includes more than a dichotomous categorization of "germline as genetic" and "tumor as genomic," which is not fully representative of all the types of genomic testing that are done in breast cancer (and other cancers as well) (Pichler et al., 2024). Genomic analysis for precision diagnosis and treatment in breast cancer is done for many purposes:

- To identify germline risk (inherited pathogen variants)
- To determine likelihood of recurrence (prognostic) and subsequent benefit from systemic adjuvant therapy (predictive) (e.g., gene expression analysis such as Oncotype DX®)
- Comprehensive germline and somatic analysis in the advanced setting (stage IV) to determine biomarker status and eligibility for targeted therapy (e.g., *PIK3CA*-activating genomic alteration and treatment with alpelisib) (National Comprehensive Cancer Network, 2024)
- To predict drug response (e.g. genotyping of *CYP2D6* alleles to determine tamoxifen metabolizer status)

The methodology in this study included that the researchers defined these terms to participants. Using consistent and accurate definitions is critical in all discussions and education about genetic/genomic terms, whether it be among healthcare professionals or patients and families. When terms are not accurately applied, the risk of error increases (Friend et al., 2021). The Oncology Nursing Society Genomics and Precision Oncology Learning Library has a taxonomy of terms that relate to genomic and precision medicine, as well as many professional resources and tools to facilitate patient education (www.ons.org/learning-libraries/precision-oncology). The umbrella term is *biomarker* and *biomarker testing* and then further separated by whether one is testing the germline or the tumor (somatic).

As Robles-Rodriguez et al. (2024) note, patients want and desire information about precision