Oncology Nursing Society Call to Action: Using the Appropriate Genomic Terminology for Safety and Quality

The Oncology Nursing Society (ONS) urges all oncology nurses to become fluent in the terms represented within the ONS Genomics Taxonomy and to use these terms in practice. The use of consistent and correct genomic terminology minimizes confusion and misconceptions and contributes to the high-quality and safe delivery of cancer care. The ONS Genomics Taxonomy serves as a foundation for terminology that will be referenced and represented across ONS materials. As the evidence base continues to evolve and expand, so will the taxonomy.

The ONS Genomics Advisory Board has developed a Genomics Taxonomy that reflects shifts in genomic terminology in accordance with the latest scientific evidence and advances in testing technologies, as well as aligns with other leading scientific organizations. Misunderstanding and incorrect use of terms can result in patient harm. Misuse of the term "mutation" has resulted in the interpretation that all mutations are detrimental, which is not accurate and can result in physical and psychological harm. For example, a malpractice case alleged that a nurse practitioner misinterpreted a genetic test result as positive when it was, instead, a variant of uncertain significance (VUS). A VUS means that there is insufficient evidence to support a more definitive classification of this variant as pathogenic or benign. This misinterpretation resulted in the nurse practitioner advising the patient to undergo surgical intervention for risk reduction (mastectomy and total abdominal hysterectomy). The patient understood her results to be positive, resulting in fear of increased cancer risk, and led her to undergo unnecessary surgical intervention (*Elisha Cooke-Moore v. Curry County Health District; Curry Community Health, Inc.; Curry Medical Practice; Curry Medical Center; Lori Johns; William Fitts; Jessica Carlson*, 2017).

A Shift in Terms

Mutation to Variant

DNA sequencing technologies have undergone rapid evolution and are able to detect multiple types of genomic variation. The functional significance of detected variants is an intense area of ongoing scientific inquiry and demonstrates the need to continually evaluate and evolve our terminology to match the scientific evidence.

The genomics community, as well as preeminent cancer organizations, such as the National Cancer Institute (NCI), have shifted from the use of "mutation" to "variant." A statement from NCI (2021b) that is aligned with the ONS Genomics Taxonomy recommendations reads,

> *A concerted effort is being made within the genetics community to shift terminology used to describe genetic variation. The shift is to use the term "variant" rather than the term "mutation" to describe a difference that exists between the person or group being studied and the reference sequence, particularly for differences that exist in the germline.*

Variants can then be further classified as benign (harmless), likely benign, of uncertain significance, likely pathogenic, or pathogenic (disease causing).

The term "mutation" is no longer appropriate to use, and its usage has led to confusion and medical errors, including those made by nurses (Friend et al., 2021).

Biomarkers and Biomarker Testing

Precise characterization of tumors through genomic analyses has led to greater understanding of the cellular, molecular, and genomic variations leading to cancer growth. Collectively, a biomarker is a biological molecule found in blood, other body fluids, or tissues that is a sign of a normal or abnormal process, or of a condition or disease. A biomarker may be used to see how well the body responds to a treatment for a disease or condition (ONS, n.d.-b).

Biomarker testing is the use of a laboratory test to measure biomarkers found in blood, other body fluids, or tissue and can be somatic or germline (ONS, n.d.-b). Biomarker discovery has led to the development of targeted treatments specific to the identified variant(s) exhibited by an individual's unique tumor. Biomarkers are a key component of precision oncology and are applicable across the cancer care continuum.

Varied terms used to describe biomarker testing (e.g., molecular profiling, tumor profiling, genomic profiling) lead to confusion and impair effective communication between patients and families and the healthcare team (NCI, 2021a).

Resources and References

Elisha Cooke-Moore v. Curry County Health District; Curry Community Health, Inc.; Curry Medical Practice; Curry Medical Center; Lori Johns; William Fitts; Jessica Carlson. (2017). http://media.oregonlive.com/pacific-northwest-news/other/mastectomy.suit.pdf

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