How Did the Variant Get Its Name? Understanding Gene and Variant Nomenclature

Patricia Friend, PhD, APRN-CNS, AOCNS®, AGN-BC, and Suzanne M. Mahon, DNS, RN, AOCN®, AGN-BC, FAAN

Genomics is foundational to precision oncology. Oncology nurses regularly review germline and somatic biomarker testing reports. The taxonomy and nomenclature of biomarker results have evolved. Accurate understanding and interpretation of germline and somatic genomic results are essential for safe patient care and patient education. This article reviews common variant nomenclature on genomic biomarker reports, including gene and variant location, coding data, information about protein function, and common DNA errors. This review includes examples of common variant types, such as insertions, deletions, duplications, and substitutions, and implications for nursing practice.

AT A GLANCE

- Germline and somatic biomarker test results include detailed information about altered genes, actionable variants, and DNA sequence and coding changes.
- Common types of DNA sequence changes include insertions, deletions, duplications, and substitution variants, which may or may not have implications for clinical care based on pathogenicity and actionability.
- Oncology nurses regularly encounter genomic biomarker reports and can explain the components and clinical implications of the reports to patients and families.

KEYWORDS
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Following the completion of the Human Genome Project in 2003, which sequenced all three billion base pairs in the human genome, international efforts moved to cataloging the DNA sequence variation among humans (Green, 2022). Humans are 99.9% identical in DNA sequence. Categorizing the 0.1% interindividual variation is the foundation for identifying what variation is associated with disease risk and disease protection (Lappalainen & MacArthur, 2021). In the genomic disease of cancer, genomic variation can occur in the germline (inherited variation) and in the tumor (somatic variation). Germline variants occur in a sperm or egg cell and are passed directly from a parent to a child. Somatic variants are acquired and happen when a gene is damaged, often from a carcinogen like tobacco.

The explosion of genomic knowledge in only two decades has revolutionized cancer care and challenges oncology nurses to keep pace with information that was never addressed in formal education programs. This includes foundational information and genomic terminology, which continue to evolve. The study of genomic variation in cancer has led to the change in terminology from "mutation" to "variant," with a qualifying term about pathogenicity, and from "single nucleotide polymorphism" to "single nucleotide variant" to describe a single nucleotide change in DNA sequence. Rapidly expanding applications of genomic variant identification and classification in cancer care have led to efforts to standardize the naming or nomenclature of variants and the reporting of pathogenicity (disease causing) and actionability (clinical utility) (Schubert et al., 2022). Oncology nurses use variant information in cancer care on a daily basis. Understanding the meaning of variant terminology and nomenclature is essential to providing safe care and effective patient and family education. This article reviews the nomenclature commonly used in germline and somatic biomarker reports as well as common types of DNA errors.

Genomic Biomarker Test Report Terminology

Biomarker test reports have extensive and specific information about the altered genes detected in an analysis. This nomenclature is presented in a standard format and provides detailed information about the variant and its location within the gene (den Dunnen, 2016). The information regarding location of a variant consists of a combination of numbers and letters in a specific order. To avoid confusion in this rapidly expanding and increasingly complex area, the Human Genome Variation Society (2020) maintains international standards for variant description (den Dunnen et al., 2016).