Confusing Genomic Terms: The Importance of Being Precise

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

Advances in genomic science directly affect cancer care. Mastery of foundational concepts is necessary to understand the rationale for many aspects of cancer care, including prevention, surveillance, and treatment of malignancy. Genomic terminology is complex and can easily be confused or even misused. Understanding genomic terms enables the oncology nurse to provide safe care and comprehensive patient and family education, which may include clarification of common misconceptions.

AT A GLANCE

- Cancer is a genomic disease, and genomic science is advancing the understanding of cancer development and enabling precision cancer care.
- Genomic science is complex and rapidly changing, and terminology can be confusing.
- Correct interpretation and application of genomic terms is critical in the provision of safe care and effective patient and family education.

KEYWORDS
genetics; genomics; genotype; phenotype; allele; variant

DIGITAL OBJECT IDENTIFIER
10.1188/24.CJON.451-456

Cancer is a genomic disease. A growing understanding of genomics as the basis for malignant disease is transforming care. Oncology nurses encounter genomic terms in clinical practice but may have educational needs regarding foundational genomic terms and taxonomy. Challenging vocabulary and conceptual complexity make learning genomics difficult (Jarvik & Evans, 2017). Although information from the Human Genome Project has been available for more than 20 years, most nursing educational programs provide little if any of this fundamental information (Seibert, 2020). Proficient and accurate application of genomic terms affects care and is essential for quality and safe patient and family education (Friend et al., 2021). This article reviews some genomic terms that can be confusing. A definition of each term is provided, followed by examples of clinical applications and implications for oncology care. An era of precision cancer care requires thorough understanding and accurate application of terminology.

Gene Versus Allele

Definitions

A *gene* is a particular region of DNA that codes for a protein that is transcriptionally active (National Human Genome Research Institute, 2024a). The human genome consists of three billion base pairs, comprising an estimated 20,000–25,000 genes. The size of a gene is determined by the number of base pairs and may range from a few hundred to more than two million. Humans are diploid. Every human has two copies of each gene, one inherited from each parent. Every gene can have different versions based on the exact sequence of DNA.

An *allele* is one particular version of a gene. Genes code for traits such as eye color or hair color, so different alleles of the gene(s) result in different exhibited traits, such as blue eyes or brown hair. Genes also influence disease risk; individuals who are born with an altered allele of the *BRCA1*/*BRCA2* tumor suppressor gene are at increased risk for a spectrum of inherited cancers.

Some genes have a limited number of alleles (possible versions), like the *ABO* gene, for which three alleles (A, B, and O) exist. In this case, the combination of two alleles inherited by an individual determines blood type (see Table 1). Other genes can have thousands of possible alleles and allele combinations. For example, the human leukocyte antigen (*HLA*) gene cluster is