Cytogenetics: Oncology Nurses in Advanced Practice
Decipher del(13q14) and t(14:16)

Stephanie Jacobson, RN, BSN, FNP-C, OCN®, Julie Eggert, PhD, GNP-BC, AOCN®, Kelly Smith, MS, FNP, BC, and Thomas Rosenzweig, MD

Advances in genetics related to the diagnosis and treatment of cancer have transformed the oncology specialty into one with more promising outcomes. Because of the Human Genome Project, the association between genetics and cancer is more clearly defined and healthcare professionals need to be prepared to integrate new genetics knowledge into clinical practice. This article reviews basic genetic information essential for oncology nurses in advanced practice. Application of genetic guidelines uses the model of multiple myeloma. Multiple myeloma is an example of a disease in which cytogenetics has become increasingly important for diagnosis, prognosis, and treatment. As the basis of knowledge in genetics continues to expand, oncology providers are transitioning to a paradigm in which cytogenetic elements carry more weight in diagnosis and treatment.

All forms of cancer are related to inherited or acquired genetic mutation. Since the launch of the Human Genome Project in 1990, the science of genetics and awareness of the role it plays in malignant transformation, growth, and treatment has grown exponentially (Human Genome Project Information, 2011). As knowledge increases about genetics and how it influences malignancies, oncology nurses in advanced practice must understand the evolving needs of patients related to new diagnosis and treatment discoveries in cancer using genetics (Jenkins, 2011).

A working knowledge in genetics assists those nurses in the identification and management of hematologic malignancies. Multiple myeloma (MM) is a malignancy of the plasma cell, affecting its ability to produce immunoglobulins, which alter the body’s ability to fight infection by producing insufficient amounts or by producing dysfunctional immunoglobulins (Tariiman & Estrella, 2005). The diagnosis of MM is confirmed with bone marrow biopsy results that detect specific genetic mutations like additions, translocations, or deletions, which may affect the prognosis. Serial biopsies monitor disease progression and evaluate treatment effectiveness. The goal of the current article is to provide genetic information that oncology nurses in advanced practice should know, using the model of MM. The synthesis analyzes cytogenetic results from bone marrow testing and teaches clinical use.

Chromosomes

Cytogenetics is concerned with the study of the structure, function, and abnormalities of chromosomes (Genetics Home Reference, 2012). Analysis includes G-banded chromosomes or other banding techniques, as well as fluorescence in situ hybridization (FISH) and comparative genomic hybridization (CGH) (Genetics Home Reference, 2012) (see Table 1). Chromosomes in the nucleus of cells contain genetic material. Each chromosomal gene codes for a specific protein (National Human Genome Research Institute, 2011a, 2011b). Proteins are essential to the structure and function of the body’s tissues and perform most of the body’s work. Scientists have estimated that each individual