

Knowledge of genetic mutations and how these affect patient outcomes is rapidly expanding in the care of individuals diagnosed with multiple myeloma (MM). Genetic analysis of bone marrow biopsies now provides information about diagnosis, response to treatment, and prognosis. Oncology nurses need to understand the science and meaning of bone marrow biopsy reports, including the implications of genetic alterations and minimal residual disease. This will allow them to provide care, support, and education related to MM and its treatment to patients and their families.

AT A GLANCE

- Cytogenetics should be considered an integral part of treatment management in patients with MM.
- Minimal residual disease testing should be performed in patients who have a complete response to treatment.
- Oncology nurses should educate themselves on gene mutations that identify a patient as having high-risk disease.

KEYWORDS

multiple myeloma; bone marrow biopsy; cytogenetics; genetics; cancer

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Multiple Myeloma

Detecting genetic changes through bone marrow biopsy and the influence on care

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Multiple myeloma, or MM, is a cancer of the plasma cells, which are found in the bone marrow and are important in immune system function. Plasma cells are responsible for producing antibodies; they respond to infection. Bone marrow analysis is a significant element in establishing the diagnosis of MM and provides necessary information on the genetic makeup of the plasma cells (Dimopoulos et al., 2011). The cause of MM remains unclear; genetic mutations are not inherited but occur during a person's lifetime. Some of these mutations play a critical role in regulating cell division, resulting in the excessive proliferation of plasma cells that distinguish MM from other diseases (Morgan, Walker, & Davies, 2012). In addition, advances in technology and the mapping of the human genome have led to discoveries that abnormalities in the expression or number of some specific genes are associated with the risk for early relapse of MM (Rajan & Rajkumar, 2015). Identifying genetic abnormalities helps providers predict the outcome of treatment for patients (see Figure 1).

Diagnosis and Response Assessment

To diagnose MM, a bone marrow biopsy must be performed (Dimopoulos et al., 2011). Biopsies are performed at specific time points throughout the disease course. This is to (a) confirm the initial diagnosis; (b) differentiate among monoclonal gammopathy of undetermined significance (MGUS), smoldering myeloma (patients

are more likely to progress to MM), and MM; (c) assess the disease at 90–100 days post-stem cell transplantation; (d) confirm remission at any time point; and (e) evaluate burden of disease and further genetic changes at each disease progression (Rajkumar et al., 2011). Despite advances in patient outcomes, more genetic markers need to be identified to improve risk prediction and to expand targeted therapy choices.

Cytogenetics, which is a technique of analyzing cells for chromosome abnormalities in MM, can be assessed by karyotype and fluorescence in situ hybridization (FISH). A karyotype is a picture of a patient's chromosomes. It can assess the appearance of the chromosomes and evaluate for deletions, additions, and translocations. FISH studies are important to help classify patients with MM into standard-, intermediate-, and high-risk groups. Patients with standard risk have the presence of chromosomal abnormalities t(11;14), t(6;14), or hyperdiploidy, whereas those with intermediate risk have the presence of t(4;14), del 13, or hypodiploidy, and those with high risk have the presence of del 17p, t(14;16), or t(14;20) (Mikhael et al., 2013). Cytogenetic abnormalities in MM influence every aspect of the disease, including progression from MGUS, a benign condition, to MM, as well as a patient's risk of relapse and his or her clinical presentation (Bergsagel, Mateos, Gutierrez, Rajkumar, & San Miguel, 2013).

Flow cytometry is a method used to examine cell surface markers, as well as the number and genetic characteristics of cells taken from bone marrow. Flow cytometry