Procedures and Interpretation of Bone Marrow Examination

Seung Yeob Lee
Department of Laboratory Medicine, Jeonbuk National University Medical School and Hospital, Korea

Bone marrow examination serves as an indispensable foundation for diagnosing, classifying, and monitoring hematologic malignancies, evolving from a strictly morphologic assessment into a highly integrated, multimodal diagnostic ecosystem. Modern hematopathology mandates seamless integration of cellular morphology, immunohistochemistry, multiparameter flow cytometry, conventional cytogenetics, and advanced molecular genetics. This comprehensive approach is necessary to meet the demands of precision medicine, in which diagnostic entities and therapeutic targets are increasingly defined by their underlying genomic architecture rather than by their visual phenotypes alone.

Ensuring the integrity of this diagnostic process begins with the rigorous standardization of preanalytical variables and specimen procurement. A standard evaluation comprises an aspirate for smear preparation and ancillary liquid diagnostics, alongside a trephine core biopsy for architectural evaluation, demanding strict adherence to specimen adequacy criteria. The exact procurement technique, including the avoidance of hemodilution and appropriate handling of dry-tap scenarios, is critical. Proper sampling preserves the statistical integrity of the myelogram and prevents false-negative flow cytometric or molecular results, ensuring that subsequent morphological and molecular evaluations are based on a truly representative sample of the marrow cavity.

Despite the rapid ascendance of genetic profiling, meticulous morphological evaluation remains an essential diagnostic anchor, particularly in complex dysplastic states and early myeloproliferative neoplasms. Deviations in the myeloid-to-erythroid ratio, lineage-specific dysplasia, and precise megakaryocyte morphological characterization are vital for differentiating benign reactive conditions from malignant clonal expansion. Furthermore, uncovering subtle bone marrow involvement in aggressive lymphomas often requires integrating morphological assessment with comprehensive immunohistochemical screening to unmask interstitial or intrasinusoidal infiltration patterns that might otherwise escape routine evaluations.

The taxonomic landscape of hematologic malignancies underwent a profound paradigm shift in 2022 with the parallel introduction of the World Health Organization 5th Edition and the International Consensus Classification. These comprehensive frameworks prioritize genomic drivers but diverge significantly in their diagnostic thresholds, specific nomenclature, and handling of intermediate disease states, most notably in their blast enumeration paradigms for acute myeloid leukemia. Navigating this complex intersection requires dual fluency to ensure accurate reporting, particularly when assessing precursor lesions, erythroid leukemias, and cases that fall within the high-risk overlap of myelodysplasia and acute leukemia.

Beyond the initial diagnosis, the evaluation of therapeutic efficacy relies on the highly sensitive detection of measurable residual disease using next-generation flow cytometry and next-generation sequencing. Achieving an MRD-negative state is a powerful prognostic indicator that directly guides consolidation and transplantation decisions. Following allogeneic hematopoietic stem cell transplantation, longitudinal chimerism analysis is required to confidently assess engraftment, rapidly identify immunological rejection, and predict impending relapse. Ultimately, synthesizing complex diagnostic data into standardized, synoptic reports ensures that critical, life-altering information is communicated unambiguously to the interprofessional hemato-oncology team.