Chapter 9
Diagnostic Approach to Myeloid Neoplasms With Predominantly Ineffective Hematopoiesis: AML, MDS, and MDS/MPNs

ABSTRACT

Acute myeloid leukemia, myelodysplastic syndromes, and myelodysplastic/myeloproliferative neoplasms share pathophysiologic features of decreased/deregulated myeloid maturation. Their diagnosis relies upon peripheral blood, bone marrow morphology, extended flow cytometry panels, detailed cytogenetic and molecular analysis tailored for each subtype, possible predisposing factors, and mimicking conditions. The overall genomic analysis provides a tool for refining risk stratification and selecting novel therapies.

INTRODUCTION

Myeloid neoplasms can manifest in various ways, including marrow failure with or without maturation arrest. Conditions like acute myeloid leukemia (AML) and myelodysplastic syndromes (MDS) fall into this category. Additionally, there are overlapping features of myeloid proliferation and defective maturation, termed Myelodysplastic/Myeloproliferative overlap syndromes (MDS/MPN).

In the patient care continuum, several critical stages and essential diagnostic tools are followed for initial diagnosis, risk stratification, response evaluation, and disease monitoring.

This chapter delves into the key clinicopathological aspects and comprehensive diagnostic approach of acute myeloid leukemia, myelodysplastic syndromes, and Myelodysplastic/Myeloproliferative neoplasms. It emphasizes the distinct diagnostic criteria, risk assessment, therapeutic strategies, and treatment response evaluation.

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ACUTE MYELOID LEUKEMIA (AML)

Acute myeloid leukemia (AML) stands as the most prevalent form of adult acute leukemia. Its incidence escalates with age, particularly in the context of myelodysplasia and therapy-related cases, with an average age of 70 years at diagnosis (Schneider et al., 2017).

AML typically manifests through marrow failure and rarely presents as a tumor mass—often solitary. This mass may serve as the initial or sole manifestation of de novo AML or a blast crisis stemming from an underlying myeloproliferative disorder, myelodysplastic syndrome, or AML relapse. Distinguishing it from a leucoerythroblastic reaction (characterized by few blasts) in myelofibrosis is of utmost importance (Bewersdorf & Abdel-Wahab, 2022).

In AML patients with hyperleukocytosis, certain emergencies warrant immediate attention. These include tumor lysis syndrome (TLS) and coagulopathy, particularly in cases like acute promyelocytic leukemia (APL). For APL, rapid treatment with all-trans retinoic acid (ATRA) is crucial—even before confirming the diagnosis and before any chemotherapy. If APL is ruled out, this treatment can be safely interrupted without adverse effects.

Clinicopathologic Features

A diagrammatic representation of AML entities defined in the ICC and WHO5 (2022) classification is shown in 1. (Rato et al., 2022).