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Opinion Article

# Impact of Complex and Monosomal Karyotypes on Outcomes in Acute Myeloid Leukemia

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## DESCRIPTION

Acute Myeloid Leukemia (AML) represents a heterogeneous group of hematologic malignancies characterized by the clonal proliferation of immature myeloid precursors in the bone marrow, leading to impaired hematopoiesis and bone marrow failure. Among the diverse biological features that define AML, cytogenetic abnormalities stand as one of the most crucial determinants of disease classification, prognosis, and therapeutic decision-making. Over the past few decades, advances in cytogenetic and molecular technologies have provided profound insights into the pathogenesis of AML, revealing that chromosomal alterations play central roles in leukemogenesis by disrupting normal gene regulation, differentiation, and apoptosis pathways. These cytogenetic aberrations, encompassing chromosomal translocations, deletions, inversions, and numerical abnormalities, not only define distinct AML subtypes but also serve as pivotal prognostic markers guiding personalized treatment approaches.

Cytogenetic abnormalities are observed in approximately 55-60% of adult AML cases, with the remaining exhibiting a normal karyotype, which itself may harbor submicroscopic molecular mutations. Among the well-established recurrent chromosomal abnormalities, certain patterns are strongly associated with favorable, intermediate, or adverse outcomes. These distinctions have become the foundation of modern AML risk stratification systems, including those developed by the European Leukemia Net (ELN) and World Health Organization (WHO). The recognition of these cytogenetic lesions has fundamentally changed the clinical management of AML, allowing oncologists to tailor therapeutic intensity, determine eligibility for hematopoietic stem cell transplantation, and predict long-term survival outcomes.

The evolution of cytogenetic analysis from conventional karyotyping to advanced molecular cytogenetic techniques, such as Fluorescence *In Situ* Hybridization (FISH), Comparative Genomic Hybridization (CGH), and Next-Generation Sequencing (NGS), has greatly expanded our understanding of chromosomal abnormalities in AML. Conventional karyotyping remains the gold standard for detecting balanced and unbalanced chromosomal changes, but its resolution is limited to abnormalities visible under a microscope. FISH, by contrast, allows for the detection of specific gene rearrangements even in metaphase-deficient samples. NGS has further revolutionized the field by uncovering cryptic structural variants, submicroscopic deletions, and single nucleotide mutations that contribute to leukemogenesis. This technological progression has bridged the gap between cytogenetics and molecular genetics, creating a more integrated approach to AML diagnosis and classification.

Therapeutic implications of cytogenetic abnormalities in AML are profound. Risk stratification based on cytogenetics determines whether a patient should receive intensive chemotherapy alone or proceed to allogeneic stem cell transplantation in the first remission. Patients with favorable cytogenetics generally benefit from standard chemotherapy, with long-term remission achievable in a significant proportion of cases. In contrast, those with adverse cytogenetics often require early transplantation, as chemotherapy alone rarely yields durable responses. The recognition of cytogenetically defined subgroups has also facilitated the development of targeted therapies aimed at specific molecular pathways. For example, FLT3 inhibitors such as midostaurin and gilteritinib have demonstrated survival benefits in FLT3-mutated AML, while IDH1 and IDH2 inhibitors have provided novel treatment options for patients harboring those respective mutations. Similarly, the identification of core-binding factor AML subtypes has led to the exploration of targeted agents that disrupt fusion protein-mediated transcriptional repression, potentially improving outcomes beyond chemotherapy.

Cytogenetic abnormalities not only serve as static prognostic markers but also offer dynamic insights into disease evolution and therapeutic resistance. Clonal evolution, whereby new cytogenetic or molecular lesions emerge during the course of disease progression or after therapy, represents a key mechanism of relapse. Monitoring these changes through serial cytogenetic and molecular testing provides valuable information for modifying treatment strategies, such as switching targeted agents, intensifying therapy or considering alternative transplant approaches. The integration of cytogenetic data with molecular profiles has led to more precise risk models, enabling the personalization of AML therapy.

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