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Chromosomal abnormalities in myelodysplastic syndromes and acute myeloid leukemia

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Abstract

Clonal chromosome abnormalities are found in more than half the patients with hematologic malignancies. Karyotype is an independent prognostic factor in these patients. Cytogenetic findings correlate significantly with morphologic, immunologic, and clinical features as well as response to treatment, remission duration, and survival. The number of different cytogenetic abnormalities is enormous; however, many cytogenetic findings frequently occur in a given disease (e.g., abnormalities of 5 or 7 in 75% to 90% of patients with therapy-related AML). Some abnormalities are found only in myeloid malignancies, for example, the t(8;21)(q22;q22) and rearrangements of chromosome 16q22, both of which have a good prognosis. Other abnormalities usually are found in both myeloid and lymphoid malignancies, for example, the t(4;11)(q21;q23) and t(9;22)(q34;q11), both of which have a poor prognosis. The Human Gene Mapping Conferences have compiled much cytogenetic data and produced several interesting correlations in myeloid malignancies: rearrangements of 3q21-26 with myeloid proliferations associated with environmental exposure (similar to abnormalities of 5q, 7q, 12p, and 17q), aberrations of 12p, 11q13 and 11q23 with both myeloid and lymphoid disorders, and the lack of myeloid involvement and abnormalities of chromosomes 14 and 18. In conclusion, cytogenetic analysis of neoplastic cells at diagnosis for patients with MDS, AML, and SAML is required for appropriate diagnosis and treatment. The use of chromosome abnormalities to separate patients into high- and low-risk groups eventually may allow us to be more effective in selecting curative therapy.

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