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Possibilities of Cytogenetic Studies in Acute Myeloid Leukemia (Literature Review)

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Abstract

The article presents a literature review of the cytogenetic aspects of diagnosis and monitoring of acute myeloid leukemia. It provides a detailed analysis of the nature of acute myeloid leukemia, its prevalence, and the possibilities of studying the morphological, immunological, and molecular genetic characteristics of blast cells. Cytogenetic methods of standard karyotyping and fluorescent in situ hybridization allow for the detection of recurrent and unique chromosomal abnormalities that lead to changes in the structure and expression of oncogenes and tumor suppressor genes, which has diagnostic and prognostic significance. The modern classification of acute myeloid leukemia is based on the morphological, immunophenotypic, and genetic features of blast cells. Identification of genetic markers - recurrent chromosomal rearrangements and gene mutations - allows for the determination of acute myeloid leukemia subtypes with favorable, unfavorable, and intermediate prognoses.

Keywords: Acute myeloid leukemia, mutation, monitoring, diagnosis, cytogenetics.

Introduction

Acute myeloid leukemia (AML) is an aggressive blood disease characterized by the rapid accumulation of immature myeloblasts in the bone marrow that suppress normal hematopoiesis, with symptoms such as anemia, bleeding, and weakness. Recent literature reviews focus on molecular genetic assessment for risk-adaptive therapy, classification by genetic anomalies (e.g., t-AML, AML-MRC), as well as new treatment approaches, including chemotherapy, to improve survival, especially in the elderly [1, 3].

AML is the most common malignant myeloid disease in adults. The incidence increases from 3.1 cases per 100,000 people aged 50-54 years to 23.1 cases among people over 80. Overall, the incidence of AML in the general population is 3.6 per 100,000. In adult patients, treatment outcomes are typically analyzed separately for young patients (18-60 years old) and elderly patients (over 60 years old). With current standard chemotherapy regimens, approximately 30-35% of adults under 60 live longer than 5 years and are considered cured. Despite significant progress in AML therapy over the past three decades, two-thirds of young people still die from this disease. Factors influencing treatment outcomes (achievement of remission, overall and disease-free survival) can be divided into those related to the patient's clinical characteristics and overall health status, and those caused by the characteristics of the tumor clone in acute myeloid leukemia [2].

Despite numerous studies conducted, in most cases, the specific cause of AML remains unknown. However, there are several predisposing factors that increase the risk of

disease: ionizing radiation, chemotherapy and radiotherapy for other tumors, smoking, prolonged exposure to benzene and its derivatives. The development of AML is based on mutations in the genetic material of the clonogenic hematopoietic cell. As a result, cell cycle control is disrupted, and the process of transcription and production of several key proteins are altered. Subsequently, due to uncontrolled proliferation and lack of differentiation, tumor (blast) cells accumulate. The detection of various chromosomal aberrations (translocations, deletions, inversions, etc.) in AML confirms that the pathogenesis of the disease is related to genetic abnormalities [5, 9].

Thus, AML is a consequence of damage (mutation) in the genetic material of a clonogenic hematopoietic cell. As a result, there is a disruption in cell cycle control, changes in the transcription process, and production of several key proteins. Due to uncontrolled proliferation in the absence of differentiation, pathological cells accumulate. The fact that the pathogenesis of AL is associated with genetic disorders is often confirmed by the detection of various chromosomal aberrations (translocations, deletions, inversions, etc.). In most cases, the specific cause of AML remains unknown. However, there are several predisposing factors that significantly increase the risk of developing this disease. The clearly proven connection between ionizing radiation from atomic bomb explosions, as well as chemotherapy and radiotherapy for other tumors, with an increased risk of AL has prompted the study of other possible leukemogenic factors (low doses of radiation, chemical substances, smoking, electromagnetic waves) [4, 8].

It has been proven that there is a dose-dependent relationship between smoking and the risk of AL development, which is especially evident for individuals over 60 years old. A number of researchers suggest that about 20% of AML cases result from smoking. Long-term exposure to benzene has a leukemogenic effect on the human body, but at low concentrations of this substance, which people most often encounter in industrial settings, no relationship with an increased risk of AML has been proven [1, 7].

Studies on the constant effects of small doses of radiation have not yet provided evidence for an increased incidence of AL. The correlation between previous chemotherapy, radiation therapy for other tumor diseases, and an increased risk of AML development was first observed in patients who had recovered from Hodgkin's lymphoma. It has been proven that not only the cumulative dose but also the intensity of the dose effect causes an increase in the incidence of AML [3, 7].

The classification of AML is based on morphological and immunophenotypic characteristics, but genetic markers are crucial for modern approaches to diagnosis and prognosis. Classification of AML by genetic markers is based on the detection of specific chromosomal rearrangements and gene mutations, which allows for the identification of AML with favorable, unfavorable, and intermediate prognoses. Key genetic markers include translocations such as t (8;21), inv (16), and t (15;17) (acute promyelocytic leukemia), as well as mutations in the NPM1, FLT3, CEBPA, DNMT3A, and IDH1/2 genes [5].

In the 2022 International Consensus Classification (ICC), the previous revised fourth edition of the WHO classification was updated, changes were made to the threshold values of blast cells and genetic categories of AML, and further expansion of the classification spectrum was carried out according to cytogenetic and mutational profiles of AML [1, 3, 6].

Cytogenetic studies in AML are necessary for making an accurate diagnosis, determining the prognosis of the disease, and choosing the most effective therapy. They reveal chromosomal abnormalities such as translocations t (15;17) in acute promyelocytic leukemia, trisomy of chromosome 8, t (8;21), inv (16) and other rearrangements that allow classifying AML and predicting disease progression [1, 6].

A cytogenetic study is conducted on the patient's bone marrow cells (karyotyping) and the material is examined to identify chromosomal changes, including rearrangements, additions, or losses of chromosomes. Acute myeloid leukemia is a rapidly progressive disease, therefore early and accurate diagnosis, including cytogenetic studies, is crucial for initiating adequate therapy and increasing chances of recovery, according to clinical recommendations of MSD Manuals and research by Hadassah Medical Moscow [6].

Cytogenetic studies such as standard cytogenetic examination (SCE) and fluorescence in situ hybridization (FISH) play a key role in diagnosing acute myeloid leukemia (AML), identifying specific chromosomal abnormalities that help classify the disease, predict its course, and choose optimal treatment strategies. SCE detects chromosomal rearrangements at the karyotype level, while FISH allows for the detection of point mutations and deletions of genes important for prognosis and response to therapy, such as FLT3, NPM1 genes, or t (15;17) associated with acute promyelocytic leukemia [3, 9].

The principle of the CCI method or karyotyping involves examining chromosomes in bone marrow cells or peripheral blood under a microscope. This method allows for the detection of large structural chromosomal abnormalities, such as translocations (transfers of segments between chromosomes), deletions (loss of segments), duplications (doubling of segments), and inversions (reversal of a chromosome segment). The significance of this method in this pathology lies in its ability to help determine the type of AML, as some chromosomal abnormalities are characteristic of certain leukemia subtypes, for example, t (15;17) in acute promyelocytic leukemia [1, 3].

Fluorescence in situ hybridization (FISH) is a molecular genetics method that uses special fluorescent probes to identify and visualize specific areas of DNA or RNA in chromosomes or cell nuclei [2, 8].

FISH allows for the detection of more subtle changes at the molecular level that are invisible in conventional

karyotyping, including point mutations, deletions, or amplifications (increase in the number of copies) of individual genes. The significance of this method in AML lies in the identification of mutations. FISH allows for the detection of specific molecular markers that have prognostic significance, such as FLT3 or NPM1 gene mutations [4].

FISH also monitors treatment responses: it can be used to determine minimal residual disease (MRD) - the presence of a small number of leukemic cells after treatment, which helps assess the effectiveness of therapy and predict recurrence. Additionally, FISH enables data confirmation and clarification, i.e., it can confirm or clarify anomalies detected using CCI, or identify other, smaller chromosomal or gene changes that are invisible in standard karyotyping.

Conclusion

Thus, acute myeloid leukemia is a malignant tumor of the myeloid blood lineage, in which altered white blood cells rapidly multiply. By accumulating in the bone marrow, they inhibit the growth of normal blood cells, which leads to a decrease in the number of erythrocytes, platelets, and normal leukocytes. This is the most common type of acute leukemia in adults, with incidence increasing with age. The five-year survival rate ranges from 15 to 70%, and remission frequency ranges from 78 to 33% depending on the disease subtype. Initially, AML is treated with chemotherapy to achieve remission; then, maintenance chemotherapy can be administered, or hematopoietic stem cell transplantation can be performed. Recent genetic studies have led to the development of tests that can accurately determine the likelihood of a patient's survival and the effectiveness of a particular drug for an individual case.

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