Analysis of Cytogenetics, Molecular Profiling, and Survival in 190 Children with Newly Diagnosed Acute Myeloid Leukemia: A Single-Center Experience

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INTRODUCTION

CHROMOSOMAL ABNORMALITIES ARE WELL-DOCUMENTED IN PAEDIATRIC ACUTE MYELOID LEUKEMIA (AML); HOWEVER, GIVEN THE RARITY OF THE DISEASE, THE PROGNOSTIC SIGNIFICANCE OF SPECIFIC CYTOGENETIC MARKERS REMAINS AN AREA OF ONGOING INVESTIGATION.

AIM

THIS STUDY AIMED TO CHARACTERIZE THE SPECTRUM OF GENETIC ABNORMALITIES IN A PAEDIATRIC AML COHORT AND TO EVALUATE THEIR ASSOCIATION WITH TREATMENT OUTCOMES.

METHOD

The study was conducted at the Scientific and Practical Medical Center for Paediatric Oncology, Haematology and Immunology (2020-2025). A total of 190 children aged ≤18 years (mean age 7.3 ± 4.9 years; 101 boys [53.2%] and 89 girls [46.8%]; M:F ratio 1.13:1) with newly diagnosed AML were enrolled. Chromosomal aberrations were identified using conventional karyotyping, FISH, and PCR. All patients received treatment according to the AML-BFM 2004 and 2023 protocols; bone marrow transplantation was not performed.

CONCLUSIONS

The findings corroborate the established prognostic relevance of the identified cytogenetic abnormalities in paediatric AML. Treatment outcomes were additionally influenced by infectious complications, nutritional status, the adequacy of intensive supportive care, and limited access to bone marrow transplantation and target therapy.

RESULTS

Karyotyping detected cytogenetic abnormalities in the form of hypo- and hyperploidy in 60% (n=114) of patients. Molecular genetic abnormalities were identified in 32.1% (n=61), with the following distribution: PML/RARA fusion in 46.0% (n=28), AML1/ETO fusion in 21.3% (n=13), inv(16) in 8.2% (n=5), MLL/AF6 fusion in 4.9% (n=3), monosomy/deletion 7 in 4.9% (n=3), KMT2A/MLLT10 fusion in 3.3% (n=2), NPM1 mutation in 6.5% (n=4), and FLT3-ITD mutation in 4.9% (n=3). Risk stratification based on genetic profile classified 23.2% (n=44) of patients as standard risk include APL children (n=28), this group treated only of combination ATO+ATRA, 66.8% (n=127) as intermediate risk, and 10.0% (n=19) as high risk. We also used sorafenib in FLT-positive patients. Complete remission rates were 98%, 89%, and 70% for standard, intermediate, and high-risk groups, respectively. Event-free survival rates were 95.5%, 44.9%, and 21.0% for the standard, intermediate, and high-risk groups, respectively. Median follow-up was 2.5 years.

Risk group	Number	%	Remission rate	EFS
standard risk	44 (28 APL)	23,2	98%	95,5
intermediate risk	127	66,8	89%	44,9(without BMT)
high-risk	19	10	70%	21(without BMT)

Genetic profile		
PML/RARA fusion	28/61	46%
AML1/ETO fusion	13/61	21,3%
inv(16)	5/61	8,2%
MLL/AF6 fusion	3/61	4,9%
KMT2A/MLLT10 fusion	2/61	3,3%
monosomy/deletion	3/61	4,9%
NPM1	4/61	6,5
FLT3-IT	3/61	4,9

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