

C13. Correlations of immunophenotypic, cytogenetic and molecular biology data in acute leukemias: Fundeni Clinical Institute Experience

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Acute leukemia represents a clinically and biologically heterogeneous malignancy with uncontrolled proliferation of hematopoietic precursors.

Materials. 90 cases of acute leukemia diagnosed in 2013 at Center of Hematology and Bone Marrow Transplantation Fundeni Clinical Institute. We have two sources of patients: Center of Hematology and Bone Marrow Transplantation and Pediatric Clinical Institute.

Methods. Analysis immunophenotyping, cytogenetic and molecular biology data in patients with acute leukemias.

Results. We analyzed 50 cases of acute lymphoblastic leukemia (ALL) and 40 cases of acute myeloid leukemia (AML) in Hematology Center of Fundeni Clinical Institute. All patients had immunophenotyping, cytogenetic examination and molecular biology at diagnosis.

From those 50 cases of ALL: 31 patients (62%) were < 18 years old and 19 patients (38%) were > 18 years old.

In patients aged <18 years old: 11 patients (35%) had pre B ALL, 15 patients (48%) had common B ALL, 4 patients (13%) had T-ALL and 1 patient was with biphenotypic ALL.

Common B ALL was associated with TEL-AML (7/50), the majority of cases showed normal karyotype and 3/50 were associated with BCR-ABL; all these patients presented at immunophenotype exam the CD66c.

Pro B ALL was associated with the MLL - AF4 (2 of the 3 patients with pro B ALL).

Patients with pre B ALL (2/11) associated E2A PBX1; 2 cases were associated TEL-AML; 3 cases showed hyperdiploidies at cytogenetic examination - the latter involving expression of CD66c.

T-ALL and biphenotypic leukemias showed no abnormalities at molecular biology except for a case that showed E2A PBX at molecular biology. In acute myeloid leukemias (without maturation, with minimal differentiation and those with maturation) no alterations were found in molecular biology exam, except of 3 cases that presented FLT3 – ITD; the last ones were associated with additional cytogenetic abnormalities. In this group, one single case of acute leukemia with maturation presented AML1 - ETO and chromosomal instability.

6/40 cases of acute myeloid leukemias showed PML- RAR α at molecular biology - one patient associated expression of CD56 and a high number of leucocytes, with unfavorable outcome.

Cases of acute leukemias with monocytic component did not associated MLL - AF9 nor the cytogenetic abnormalities and only 3 of these patients presented NG2; one case of acute myelomonocytic leukemia presented CBFB - MYH11 type A; also 5 cases associated FLT3 – ITD.

Conclusions. In all cases, the immunophenotype diagnosis was correlated with molecular abnormalities, according to the literature data. CD66c expression was found in all patients diagnosed with hyperdiploidies and in cases of acute lymphoblastic leukemia with BCR- ABL. MLL - AF9 mutation was not found in any case of acute monocytic leukemia. NG2 expression in acute leukemias was associated with monocytic component, but with a low frequency.