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Original Research Article

Clinical, immunophenotypic and cytogenetic profile of acute lymphoblastic leukemia in children aged 1–12 years at a tertiary care centre in central India: a prospective observational study

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ABSTRACT

Background: Acute leukemias, particularly ALL, constitute one of the most common and curable malignancies in children across India. Their heterogeneity mandates detailed evaluation of clinical, immunophenotypic and cytogenetic features to guide therapy and prognostication. Indian studies highlight variable survival, with rural populations often disadvantaged in access and outcomes.
Methods: We conducted a two-year prospective study (2022–2024) at a tertiary center in central India to assess the clinical, immunophenotypic and cytogenetic profile of children (1–12 years) with newly diagnosed ALL. The study compared clinical features, immunophenotypic subtypes and cytogenetic abnormalities between patients with poor outcomes and others to improve diagnostic accuracy, risk stratification and treatment planning in resource-limited settings.
Results: This study evaluated 40 pediatric patients with ALL to identify factors influencing clinical outcomes. B-cell ALL was the predominant subtype (82.5%) and was associated with better outcomes compared to T-cell ALL. Extremes of age (<3 years and >10 years), high total leukocyte count (>50,000/µl), severe thrombocytopenia and high-risk stratification were significantly associated with poorer outcomes. Immunophenotypic markers suggestive of T-cell lineage and unfavourable cytogenetic abnormalities such as t (9;22) were linked to poorer prognosis, while t (12;21) was associated with favorable outcomes. Overall, only 40% of patients achieved remission, highlighting the need for early risk stratification and close monitoring of high-risk children.
Conclusions: Immunophenotypic and cytogenetic profiles significantly influenced patient outcomes. Specifically, T-cell markers (CD3, CD5, CD7) and unfavorable translocations like t (9;22) or t (1;19) were associated with a poor prognosis. Conversely, B-cell markers (CD19, CD10) and the t (12;21) translocation predicted more favorable results. Although the cohort was stratified into standard (22.5%), intermediate (65%) and high-risk (22.5%) groups, the trend linking high-risk status to poorer outcomes was not statistically significant, likely due to the small sample size.

Keywords: Acute lymphoblastic leukemia, Cytogenetic profile, Immunophenotypic, Pediatric oncology

INTRODUCTION

ALL is the leading pediatric malignancy, accounting for approximately 25–30% of childhood cancers.[1-3] Characterized by the clonal proliferation of immature lymphoid precursors, ALL is biologically diverse, categorized primarily into B-cell (more common, better prognosis) and T-cell subtypes (more aggressive).[1-3] While high-income countries have achieved survival rates exceeding 80% through risk-adapted chemotherapy, outcomes in low- and middle-income regions like India remain inconsistent due to resource limitations and

delayed diagnoses.[4,5] Consequently, precise diagnostic tools-including immunophenotyping and cytogenetic analysis-are critical for tailoring treatments and improving global survival rates.[6,7]

Key demographic and clinical factors serve as vital predictors for childhood ALL outcomes and form the foundation for risk-adapted therapy. Age at diagnosis is a primary indicator; children aged 1–10 years typically experience better survival rates than infants or adolescents.[8] Conversely, factors such as male gender, high initial leukocyte counts, bulky disease, central nervous system involvement and severe cytopenias are consistently associated with a poorer prognosis and require more intensive treatment strategies.[6,8]

Cytogenetic and molecular abnormalities play a crucial role in determining prognosis and therapeutic response in pediatric ALL.[7,10,11] Favorable cytogenetic abnormalities such as t (12;21) (ETV6–RUNX1) and hyperdiploidy are associated with improved survival, whereas unfavourable abnormalities including t (9;22) (BCR–ABL1), t (4;11) and t (1;19) are linked to increased risk of relapse and treatment failure.[8,11] Incorporation of cytogenetic data into treatment protocols has significantly enhanced risk-adapted therapy.[7]

In resource-limited settings, additional challenges such as delayed presentation, increased infection-related morbidity, socioeconomic barriers and treatment abandonment further contribute to poorer outcomes.[4,5] Understanding the demographic, clinical, immunophenotypic and cytogenetic factors influencing prognosis in such settings is essential to improve early risk stratification and optimize treatment strategies.[4,5] The present study was undertaken to analyse the demographic profile, clinical features, laboratory parameters, immunophenotypic and cytogenetic characteristics of children diagnosed with acute lymphoblastic leukemia and to correlate these factors with treatment outcomes. Identifying predictors of poor prognosis may aid in early risk assessment, individualized treatment planning and ultimately improve survival outcomes in pediatric ALL.[6,7,18]

METHODS

Study design and setting

This prospective observational study was conducted in the Department of Pediatrics, Government Medical College, Nagpur, over 24 months (January 2022–January 2025). Ethical clearance was obtained from the Institutional Ethics Committee and written informed consent was taken from all guardians.

Study population

Children aged between 1-12 years, who are diagnosed cases of ALL.

Sample size and sampling

The sample size is based on clinical profile of cases reported in published article by Guna Pandian & Murugesa Sankarasubramanian from the clinical signs in acute leukemia patients at presentation and is calculated using software n Master 2.0 with expected proportion of 77.4 %, precision of 15, desired confidence interval (1-alpha) 95 % and sample size was calculated to be 30.

Data collection and procedures

The study was conducted as a prospective observational investigation at a tertiary care center in central India from 2022 to 2024, after obtaining approval from the Institutional Ethics Committee and written informed consent from parents or guardians. The population included children aged 1–12 years newly diagnosed with ALL, with participants recruited consecutively based on inclusion and exclusion criteria. Patients who had received prior chemotherapy elsewhere or lacked consent were excluded from the analysis.

A detailed history and physical examination were performed for each participant, followed by laboratory investigations. Bone marrow aspiration was performed to confirm the diagnosis and immunophenotyping via flow cytometry determined lineage (B-cell or T- cell), while cytogenetic analysis by RT-PCR and gel electrophoresis identified major translocations such as BCR-ABL, ETV6-RUNX1 and E2A-PBX1. Risk stratification was carried out according to national cancer institute (NCI) criteria, which considered age at diagnosis, initial leukocyte count, cytogenetic profile and extramedullary involvement.

All patients received chemotherapy protocols tailored to their risk stratification group. Minimal residual disease (MRD) was assessed after induction and consolidation therapy. Outcome measures included remission status, induction failure, relapse, abandonment and death. Data collection procedures and follow-up visits were standardised and efforts were made to minimize bias by applying identical diagnostic and treatment protocols to all participants. Data analysis included appropriate statistical methods to evaluate predictors of outcome and control for confounding variables.

Statistical analysis

Data were entered into Microsoft Excel and tables and graphs were prepared using Excel and Word. Continuous variables were summarised as mean±SD or as median with range for non- normal data, while categorical variables were expressed as frequencies and percentages. Demographic variables were compared between groups using the chi-square test, with Fisher's exact test applied where numbers were small. Independent t-tests were used to compare pre- induction haemoglobin and peripheral smear blasts with outcomes, while non-normal variables

such as TLC and platelet counts were analysed using the Mann–Whitney test. A p value<0.05 was considered statistically significant. All analyses were performed using STATA version 14.0.

RESULTS

As per Figure 1, in this prospective observational study evaluating children aged 1–12 years diagnosed with ALL and treated under the COG ALL protocol, immunophenotypic classification demonstrated a predominance of B-cell lineage ALL. Out of a total of 40 enrolled cases, 33 patients (82.5%) were diagnosed with B-cell ALL, while 7 patients (17.5%) had T-cell ALL. Immunophenotyping was performed using flow cytometry based on specific surface markers, in accordance with standard diagnostic guidelines. The higher frequency of B-cell ALL observed in this study is consistent with the established epidemiological pattern seen in pediatric ALL populations.

As per Table 1, in this prospective observational study, the correlation between age at diagnosis and treatment outcome was evaluated among children with ALL. The age-wise distribution showed that the majority of patients belonged to the 3-6 years age group (21 cases, 52.5%), followed by 7-10 years (12 cases, 30%), >10 years (5 cases, 12.5%) and <3 years (2 cases, 5%). Analysis of outcomes revealed that poorer outcomes were more frequently observed at the extremes of age. Among children aged less than 3 years, 6 out of 8 patients (75%) had poor outcomes, while in those older than 10 years, 5 out of 7 patients (71.4%) showed poor outcomes. In contrast, children in the intermediate age groups had relatively better outcomes, with only 3 out of 15 patients (20%) aged 3-6 years and 1 out of 10 patients (10%) aged 7-10 years experiencing poor outcomes. Overall, the findings suggest that although the highest number of cases occurred in the 3-6 years age group, the risk of adverse outcomes was higher among younger (<3 years) and older (>10 years) children.

As shown in Table 2, fever was the most common presenting symptom among children with acute lymphoblastic leukemia, observed in 30 patients (75%). This was followed by anorexia and weight loss in 20 patients (50%). Easy fatigability and neck swelling were each reported in 17 patients (42.5%). Abdominal distension and cough/breathlessness were less frequent, each occurring in 10 patients (25%). Overall, systemic symptoms such as fever, fatigue and weight loss predominated, highlighting their importance as early clinical indicators in children with acute lymphoblastic leukemia. As per Table 3, The immunophenotypic analysis showed that CD19 (24 cases) and CD10 (22 cases) were the most commonly expressed markers, typical of B-cell ALL. This profile helps in classifying the type of ALL and is crucial for determining treatment protocols. CD3 and CD7, markers associated with T-cell ALL, were less frequently observed, consistent with the lower number of T-cell ALL cases in this cohort. Poor outcomes were more common in patients expressing CD3, CD5 and CD7, which are typically associated with T-cell ALL. In contrast, B-cell markers such as CD19 and CD10 were linked to better outcomes. This highlights the prognostic significance of immunophenotyping, as T-cell ALL markers are often associated with a more aggressive disease and poorer prognosis.

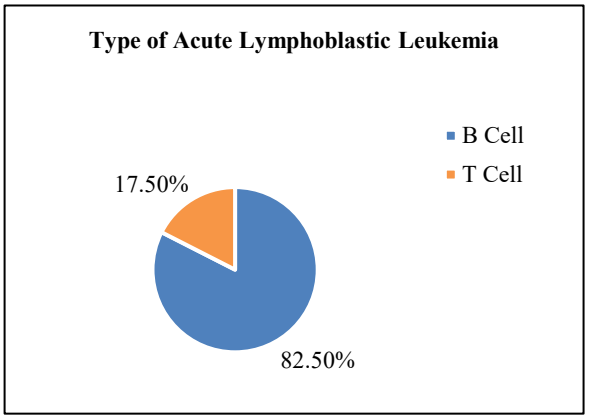

Figure 1: Distribution of immunophenotype of leukemia.

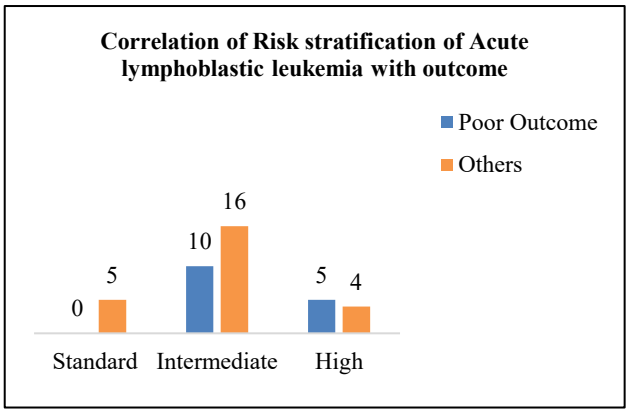

Figure 2: Correlation of risk stratification of acute lymphoblastic leukemia with outcome.

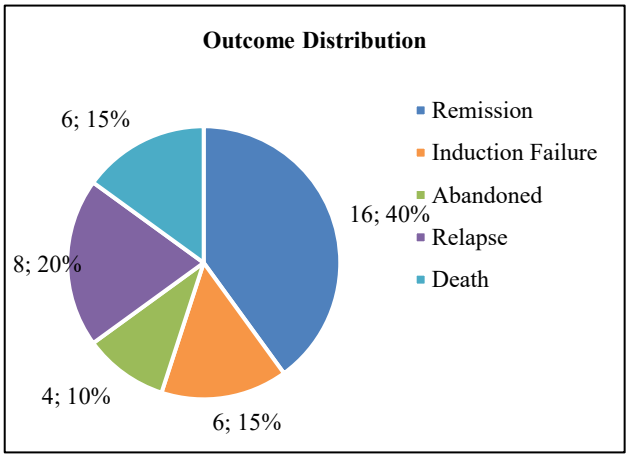

Figure 3: Outcome distribution.

As per Table 4, cytogenetic abnormalities such as t (12;21), associated with a favorable prognosis, were found in 6 cases, while t (9;22), linked to a poor prognosis, was found in 3 cases. These findings underscore the importance of cytogenetic analysis in predicting outcomes, as specific chromosomal translocations are strongly associated with prognosis and guide treatment decisions. As illustrated in Figure 2, the correlation between risk stratification and outcome in children with acute lymphoblastic leukemia shows a clear variation across different risk groups. Among the standard-risk group, none of the patients had a poor outcome, while all 5 patients were categorized under other outcomes. In the intermediate-risk group, 10 patients experienced poor outcomes, whereas a higher number, 16 patients, had other outcomes. In contrast, the high-risk group showed a relatively greater proportion of poor outcomes, with 5 patients affected compared to 4 patients with other outcomes. These findings suggest that higher risk stratification is associated with an increased likelihood of poor outcomes. As depicted in Figure 3, the overall outcome distribution among the study population demonstrates that remission was the most common outcome. However, a significant proportion of patients experienced adverse outcomes, including relapse, induction failure, abandonment of treatment and death. Among these, relapse and death contributed notably to unfavorable outcomes, while induction failure and treatment abandonment were observed in a smaller proportion of cases. This distribution highlights the variability in treatment response and underscores the need for early risk stratification and close monitoring to improve overall prognosis in children with acute lymphoblastic leukemia.

Table 1: Correlation of age in years with outcome.

Age wise distribution of cases of acute lymphoblastic leukemia		
Age (in years)	**Number of patients**	**%**
<3	2	5
3–6	21	52.5
7–10	12	30
>10	5	12.5

Table 2: Presenting symptoms of acute lymphoblastic leukemia.

Presenting symptoms of acute lymphoblastic leukemia		
Symptom	**Number of patients**	**%**
Fever grade	30	75
Abdominal distension	10	25
Easy fatiguability	17	42.5
Anorexia/weight loss	20	50
Neck swelling	17	42.5
Cough/breathlessness	10	25

Table 3: Correlation of immunophenotypic profile with poor outcome in acute lymphoblastic leukemia.

Correlation of immunophenotypic profile with poor outcome in acute lymphoblastic leukemia		
	Poor outcome	**Others**
CD3	6	2
CD5	4	0
CD7	4	2
CYCD3	4	0
CD19	14	10
CD10	12	10
CD20	4	1
CD22	7	2

Table 4: Cytogenetic profile of acute lymphoblastic leukemia in children.

Cytogenetic profile of acute lymphoblastic leukemia in children		
Cytogenetic profile	**Number of patients**	**%**
t (9, 22) (q34, q11)	3	7.5
t (12, 21) (p13, q22)	6	15
t (1, 19) (q23, p13)	3	7.5
t (4, 11) (q21, q23)	0	-

DISCUSSION

ALL is a biologically heterogeneous disease and its outcome is influenced by a complex interplay of demographic, clinical, immunophenotypic, cytogenetic and treatment-response–related factors. The present study evaluated 40 children with ALL and analyzed these parameters in relation to treatment outcomes, thereby providing insight into prognostic determinants in a resource-limited setting. Similar comprehensive evaluations have been emphasized by Pui et al who highlighted the multifactorial determinants of prognosis in pediatric ALL.[6,9] In this study, B-cell ALL constituted the majority of cases (82.5%), while T-cell ALL accounted for 17.5%, which is consistent with previously published Indian and international studies. A comparable distribution was reported by Pandian et al who also observed B-cell lineage predominance in pediatric ALL.[13] Patients with B-cell ALL demonstrated relatively better outcomes, whereas T-cell ALL was associated with a higher proportion of poor outcomes, supporting findings by Hunger and Mullighan, who described T-cell ALL as having more aggressive disease biology.[7]

Ba-Saddik et al from Yemen reported leukemia as the leading childhood cancer, forming the majority of cases, although detailed immunophenotypic classification was limited.[19] In Iran, Mousavi et al similarly identified ALL as the most frequent paediatric malignancy and noted that B-cell ALL constituted the predominant subtype, aligning with global trends.[20] Data from Jordan by Al-Sheyyab et al also showed leukemia as the most common cancer in children, with ALL being the major subtype, though lineage-specific distribution was not extensively detailed.[21] In Jamaica, Bishop et al reported a high burden of leukemias among childhood cancers, with ALL forming a significant proportion, consistent with patterns seen in other populations.[22]

Age at diagnosis emerged as an important prognostic factor, with children younger than 3 years and older than 10 years showing poorer outcomes compared to those between 3 and 10 years. This observation is in agreement with the study by Smith et al which demonstrated superior survival in children aged 1–10 years.[8] Similarly, Lee et al reported poorer outcomes at age extremes, while Siddaiahgari et al noted a higher incidence in younger children, indicating demographic variability.[14,15] Risk stratification played a crucial role in predicting outcomes in this study. Patients were categorized into standard-risk (5 cases), intermediate-risk (26 cases) and high-risk (9 cases), with intermediate-risk forming the majority. Poor outcomes were predominantly observed in the high-risk group, while none were seen in the standard-risk group. Similar trends were reported by Nachman et al who demonstrated that outcomes worsen with increasing risk category, validating the role of risk-adapted therapy.[11]

Immunophenotypic analysis in this study highlighted its important role in prognostication. CD19 (80%) and CD10 (50%) were the most frequently expressed markers and their expression, along with CD20, CD22, CD3 and CD5, showed a significant association with poor outcomes. While B-cell markers are traditionally considered favorable, their association with adverse outcomes in this cohort suggests disease heterogeneity. Similar observations were reported by Pandian et al who also noted that additional markers such as CD34 and MDR1 may influence prognosis.[16] A significant association was found between the expression of CD19 ($p=0.028$) and CD10 ($p=0.014$) with poor outcomes, including relapse, induction failure and death, suggesting a possible link with higher leukemic burden or more aggressive disease biology. Additionally, expression of CD20 ($p=0.036$), CD22 ($p=0.008$), CD3 ($p=0.036$) and CD5 ($p=0.015$) also demonstrated significant correlation with adverse outcomes. In contrast, CD7 expression did not show a statistically significant association ($p=0.174$), indicating limited prognostic relevance in this cohort.

While traditionally T-cell markers such as CD3 and CD5 are associated with poorer prognosis and B-cell markers like CD19 and CD10 with better outcomes, the findings of this study suggest variability in prognostic implications, thereby reinforcing the importance of comprehensive immunophenotypic evaluation for risk stratification and therapeutic planning in pediatric ALL. Cytogenetic analysis demonstrated a significant impact on outcomes. BCR-ABL1 positivity (t (9;22)) was significantly associated with poor outcomes ($p=0.020$), consistent with findings by Moorman et al who identified it as a high-risk abnormality.[12] Favorable cytogenetics such as t (12;21) were associated with better outcomes, whereas t (1;19) and t (9;22) were linked to poor prognosis. These findings align with studies by Harrison et al which emphasized the prognostic importance of cytogenetic abnormalities in ALL.[12]

The overall remission rate in this study was lower compared to high-income countries, with a notable proportion of patients experiencing relapse, induction failure, abandonment or death. This is comparable to findings by Asthana et al who highlighted the challenges in resource-limited settings, including delayed diagnosis, infections and treatment non-compliance affecting outcomes.[18] Overall, this study reaffirms that B-cell ALL is the predominant subtype in Indian children, with intermediate-risk disease forming the majority. Poor prognostic factors identified include T-cell lineage, high leukocyte count, low hemoglobin, adverse cytogenetics and MRD positivity. These findings are consistent with those reported by Inaba et al who emphasized the integration of clinical and biological factors for improved risk stratification and outcomes in pediatric ALL.[4]

Strengths

The strength of this study lies in its comprehensive evaluation of demographic, clinical, immunophenotypic, cytogenetic and MRD parameters in relation to outcome.

Limitations

However, limitations include the relatively small sample size and single-center design, which may limit generalizability. Larger, multicenter studies are needed to further validate these findings.

CONCLUSION

In conclusion, this study highlights the complex nature of pediatric ALL and shows how clinical features, immunophenotypic patterns and cytogenetic abnormalities strongly influence prognosis and treatment response. An overall remission rate of 85% was achieved, while 15% of children experienced induction failure. Risk stratification helps in identifying the high risk patients and sorting the treatment accordingly. Those patients with high risk categories showed poor prognosis and outcomes. Poorer outcomes were most often linked to MRD levels above 0.01%, high leukocyte counts at diagnosis and high- risk cytogenetic changes such as the BCR-ABL fusion gene. Incorporating cytogenetics and immunophenotyping in risk stratification particularly helped to identify the patients with high and intermediate risk and to intensify the treatment modality.

The study further emphasizes the value of personalized approaches-combining advanced diagnostics like flow cytometry and molecular testing with ongoing MRD monitoring to optimize outcomes. Such strategies could improve long-term remission, lower relapse rates and ultimately enhance survival in children with ALL. Moving forward, research should aim to develop newer therapies for high-risk subtypes and strengthen early detection tools, with the broader goal of reducing treatment resistance and improving both survival and quality of life for affected children.

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