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# Pediatric B-Lymphoblastic Leukemia with Aberrant CD117 Expression

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## Abstract

CD117 (c-Kit) is a tyrosine kinase receptor primarily expressed in myeloid lineage cells and certain acute myeloid leukemia (AML) subtypes. Its aberrant expression in acute lymphoblastic leukemia (ALL), particularly B-lymphoblastic leukemia (B-ALL), is rare. We present a pediatric case of B-ALL with unusual CD117 positivity and review of relevant literature to assess its prevalence, diagnostic challenge and clinical implications.

An 8-year-old girl presented with prolonged fever and melena, pancytopenia and 12% blasts on peripheral smear. Bone marrow immunophenotyping revealed 90% blasts expressing B-cell markers alongside myeloid markers CD117 and CD33, but negative for myeloperoxidase and monocytic markers, excluding mixed phenotype acute leukemia. Cytogenetics showed ETV6::RUNX1 fusion and hypertriploidy. The patient was treated with standard induction chemotherapy but developed fatal neutropenic colitis complicated by fungal infection.

CD117 expression in B-ALL is uncommon and may complicate diagnosis. Comprehensive immunophenotyping and molecular analysis are critical for accurate classification. While CD33 is frequently associated with ETV6::RUNX1-positive B-ALL, the prognostic role of CD117 remains unclear, warranting further study.

Key words: CD117 aberrant expression, BALL with ETV6::RUNX1 fusion, pediatric BALL.

## Introduction

CD117 (c-Kit) is a transmembrane tyrosine kinase receptor integral to the regulation of hematopoietic stem cell survival, proliferation and differentiation.[1] Its expression is predominantly associated with cells of the myeloid lineage and is characteristically observed in certain subtypes of acute myeloid leukemia (AML), including patients with CEBPA mutations.[3] Aberrant expression of CD117 in acute lymphoblastic leukemia (ALL), however, has been infrequently documented.[2] We report a case of pediatric B-lymphoblastic leukemia (B-ALL) exhibiting CD117 positivity. This report is accompanied by a review of the current literature to discuss the prevalence, diagnostic challenge and potential clinical implications of CD117 expression in B-ALL.

## Case Report

An 8-years-old female patient, presented with fever for 3 months and melena for 1 week. Physical examination revealed distended abdomen, rest of the systemic examinations were unremarkable. Her vitals were; BP 130/90 mm Hg, pulse 114 beats per minute, respiratory rate 30 per minute and temperature 37 ℃. Weight of the patient was 14 kg. Birth and family history is unremarkable. Immunizations are up to date. Three months back patient was admitted at another facility, where she received antibiotics for fever and red cells transfusion due to anemia. In-house CBC reported pancytopenia; Hb 6.3 g/dl, WBC count 6.5 x 10^9/L and Platelet count 30 x 10^9/L. On differential leucocyte count, 12% blast cells were seen. Rest of the cells comprised of 1% neutrophils and 87% lymphocytes. Blood and urine cultures showed no growth. Ultrasound abdominal revealed mild ascites. Bone marrow biopsy was then performed for leukemia workup. Immunophenotyping by flow cytometry (Figure 1} of bone marrow aspirate demonstrated a predominant population (90%) of small to medium-sized blasts.

These cells exhibited a characteristic B-lymphoid immunophenotype, expressing terminal deoxynucleotidyl transferase (TdT), CD19, CD79a, CD22, CD10, CD38, CD58, and dim CD45. Additionally, the blasts displayed heterogeneous expression of CD9 and CD73, alongside notable positivity for CD123. Interestingly, aberrant expression of myeloid-associated markers CD117 and CD33 was also observed.

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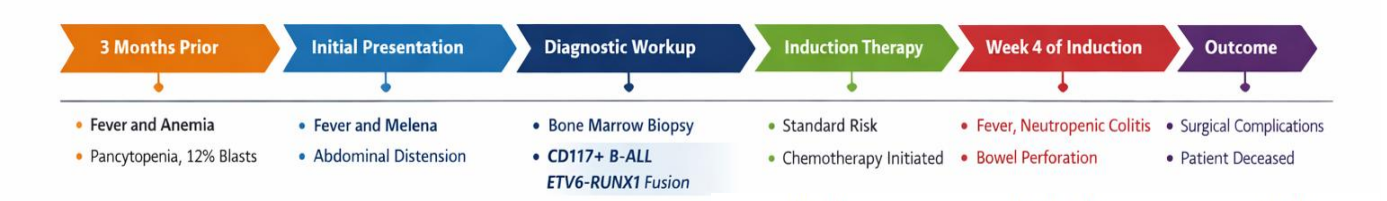


Figure 1. Timeline of the patient progress, from initial presentation, diagnosis to outcome.

The blasts were uniformly negative for CD34, CD20, myeloperoxidase (MPO), and monocytic markers including CD36, CD61, CD64, CD11b, CD11c, as well as for intracellular CD3 and Glycophorin A. According to the 2022 WHO Classification criteria for mixed phenotype acute leukemia (MPAL), confirmation of myeloid lineage requires MPO and/or monocytic marker expression, which was absent in this case. Consequently, despite the aberrant expression of CD117, the immunophenotypic profile was consistent with B-Lymphoblastic Leukemia (B-ALL). In-house available FISH probes for BALL and AML (Figure 2) revealed positivity for ETV6::RUNX1 fusion gene, while it was negative for BCR::ABL1 fusion gene, RUNX1::RUNX1T1 fusion gene, KMT2A and CBFB gene rearrangements. Moreover, extra signals were seen in all the probes tested. Karyotype (see Figure 3) revealed an abnormal hypertriploid female chromosome complement (ISCN: 78<3n>,XX,-X,+2,+4,+6,-7,+10,+13,+16,+17,+17,+18,-19,+21,+21,+22[2]/46,XY[18]).

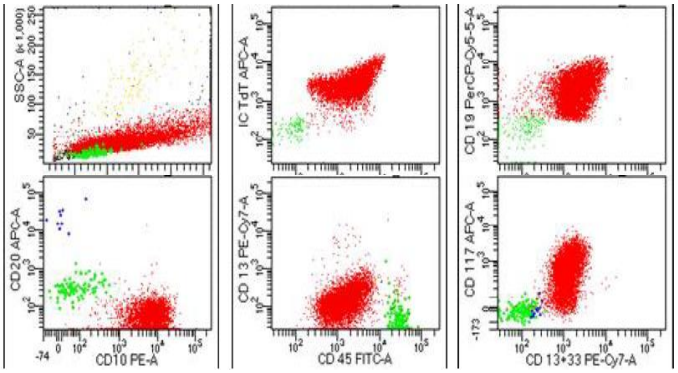


Figure 2. Flow cytometric analysis dot plots. Red coded population is blasts. It is positive for CD79a, TdT, CD22, CD19, CD10, CD33. It is heterogenous positive for CD117 while dim positive for CD45. Green coded population is benign T-cells.

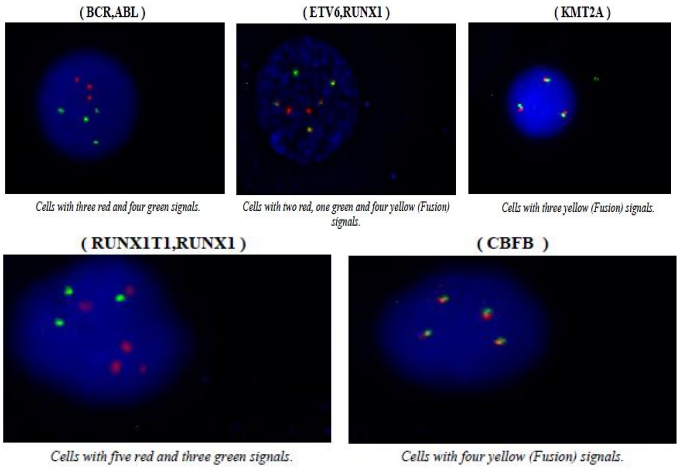


Figure 3. FISH analysis. It reported positive for ETV6: RUNX1 fusion gene while negative for BCR::ABL fusion gene, RUNX1::RUNX1T1 fusion gene, KMT2A and CBFB gene rearrangements. However, all the probes show gain of signals.

Figure 4. Karyotype. It revealed abnormal hypertriploid female chromosome complement. ISCN: 78<3n>,XX,-X,+2,+4,+6,-7,+10,+13,+16,+17,+17,+18,-19,+21,+21,+22[2]/46,XY[18].

The patient was stratified as standard risk and induction chemotherapy initiated, with good response to prednisolone prophase. In the last week of induction, patient developed fever and presented in ER. She was lethargic, tachypneic and there was abdominal distention as well as mucositis. CBC reported Hb 7.1 g/dl, WBC count 0.84 x 10^9/L and PLT count 5 x 10^9/L. Absolute neutrophil count was 0.27 x 10^9/L. Blood culture

showed growth of *E. coli*. CT scan findings were suggestive of bowel perforations. She was admitted under the impression of neutropenic colitis and started on IV medications. She also received blood transfusions. Exploratory laparotomy was performed and histopathological findings showed inflamed granulation tissue with fungal infection in duodenum and jejunum. The patient unfortunately couldn't recover and later expired.

## Discussion

The aberrant expression of CD117 in B-ALL raises important diagnostic considerations, as it may mimic features of mixed phenotype acute leukemia (MPAL) or acute myeloid leukemia (AML). However, as per the 2022 WHO Classification, MPAL requires expression of definitive myeloid markers such as myeloperoxidase (MPO) or monocytic markers (CD11b, CD64, CD36), which were absent in our case.[3] This distinction is critical for accurate diagnosis and treatment planning, as B-ALL and MPAL require different therapeutic approaches and carry different prognostic implications.[2,5] While, CD33 expression has frequently been observed in ETV6::RUNX1 fusion gene[3], CD117 expression is rare and hence its prognostic significance is yet to be studied.[6]

## Conclusion

This case highlights the rare occurrence of CD117 expression in pediatric B-lymphoblastic leukemia, emphasizing the importance of thorough immunophenotypic and molecular evaluation to avoid diagnostic pitfalls. While aberrant CD117 and CD33 expression in B-ALL may not indicate mixed lineage, their presence warrants careful interpretation within the context of established diagnostic criteria. Further research is needed to elucidate the clinical and prognostic significance of CD117 in B-ALL, which may ultimately inform risk stratification and therapeutic approaches.

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