Genetic Mutations Detected in Adult Patients Diagnosed With Acute Lymphoblastic Leukemia in a Public Hospital in Guatemala, From 2022 to May 2025

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Introduction

The global trends on genetic mutations, treatment response, and prognostics on patients with acute lymphoblastic leukemia (ALL) are relevant to providing the best quality of life to every patient and to allocate medical funds and resources appropriately. (1,2)
The detection of molecular abnormalities has implied a hallmark in its treatment, as these directed therapeutics have improved the results in certain groups, as patients positive to t(9;22) *BCR::ABL1*. (3)
To this date, there is no data regarding the genetic mutations present in Guatemalan patients diagnosed with ALL.

Aim

To describe the genetic mutations detected in patients diagnosed with ALL in the Hematology Unit in Hospital Roosevelt from 2022 to May 2025.

Objectives

- To describe the demographic and diagnostic characteristics diagnosed with ALL.
- To determine the percentage of patients positive to *BCR::ABL1, KMT2A::AFF1, TCF3::PBX1,* and *ETV6::RUNX1* mutations.

Methods

Descriptive, retrospective, cross-sectional, observational analysis from the data of 94 patients diagnosed with ALL in a public tertiary-level hospital in Guatemala from 2022 to May 2025. The diagnosis of ALL was confirmed through flow cytometry, bone marrow biopsy, or bone marrow morphology. Four genetic variants were analyzed on bone marrow or peripheral blood using RT-PCR: t(9;22) *BCR::ABL1*, t(12;21) *ETV6::RUNX1*, t(4;11) *KMT2A::AFF1*, and t(1;19) *TCF3::PBX1*. Patients with inconclusive immunophenotyping were excluded (one case). The information was collected from the medical records from the Hematology Unit and analyzed using Microsoft Office Excel.

Results

Table 1. Demographic and diagnostic characteristics of patients with ALL, from 2022 to May 2025

Median age (range) - years	33 (18 - 65)
Sex - no. (%)	
Female	46 (48.94%)
Male	48 (51.06%)
Cases per year - no. (%)	
2022	18 (19.15%)
2023	25 (26.60%)
2024	35 (37.23%)
May 2025	16 (17.02%)
Diagnosis - no. (%)	
ALL	3 (3.19%)
FAB ALL L1	6 (6.38%)
FAB ALL L2	13 (13.83%)
FAB ALL L3	3 (1.06%)
B-cell ALL	38 (40.43%)
Pre-B ALL	28 (29.78%)
Pro-B ALL	1 (1.06%)
T-cell ALL	1 (1.06%)
Acute leukemia mixed phenotype, B/Myeloid	2 (2.17%)
Acute leukemia mixed phenotype, rare types (B/T)	1 (1.06%)

Figure 1. Results of cualitative genetic analysis of patients diagnosed with ALL, from 2022 to May 2025

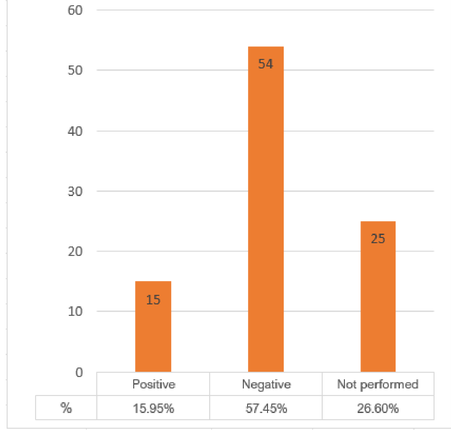
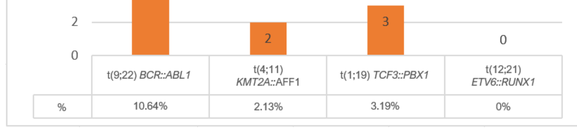


Figure 2. Genetic mutations detected in patients diagnosed with ALL, from 2022 to May 2025



Summary

A genetic mutation was detected in 15.95% of adults with ALL. The most frequent genetic mutation detected was t(9;22) *BCR-ABL1* in 10.64% of cases.

Discussion

The higher presence of *BCR::ABL1* in this study correlates with the data published worldwide where this genetic mutation is found to be the most frequently observed in adult populations. (3,4)

It highlights the importance of their measurement, as this helps the clinician to make better individual therapeutic decisions.

The percentage of patients where mutational analysis was not performed reflect limited RT-PCR availability, restricted sample-shipping days/schedule, and difficulty obtaining enough bone marrow samples.

A larger study with a sociodemographic emphasis is required to determine more robust mutational tendencies in the Guatemalan population.

References

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