Mutational Status of IGHV and Common Cytogenetic Abnormalities in Newly Diagnosed Patients With Chronic Lymphocytic Leukemia in a Hospital in Guatemala City From 2023 to April 2025

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Introduction

Chronic lymphocytic leukemia (CLL) is the most common type of leukemia diagnosed. Its incidence is higher in older patients, and its clinical course is highly variable. (1) The disease is defined by a mature clonal B-Cell lymphocyte population 5≥10^9/L in peripheral blood for more than 3 months. (2). The diagnosis is made through various methods that include immunophenotype which identify a clonal B-Cell population carrying the CD5 antigen as well as typical B-Cell markers. (1). The mutational status of Immunoglobulin Heavy Chain Variable (IGHV) region is a significant prognostic factor. Mutated IGHV is associated with better prognosis and longer survival. Around 80% of all patients with CLL carry at least one of four common cytogenetic alterations: a deletion in chromosome 13q14.3 (del(13q)), del(11q), del(17p), or trisomy 12. Del(13q) is the most common chromosomal alteration, occurring in about 55% of all cases. An isolated del(13q14) usually indicates a less aggressive form of the disease. Mutations of *TP53* are found in 4%–37% of patients with CLL and are associated with inferior prognosis. Trisomy 12 is observed in 10%–20% of patients with CLL and is associated with an intermediate prognosis (3).

Objective

To describe the mutational status of IGHV and incidence of common cytogenetic abnormalities in patients with newly diagnosed CLL from 2023 to April 2025.

Methods

Retrospective observational analysis from 2023 to April 2025. Data was retrieved from 27 patients with newly diagnosed CLL who underwent Next generation sequencing (NGS) or Fluorescent in situ Hybridization (FISH) study for cytogenetic alterations and polymerase chain reaction (PCR) for mutational status of IGHV in Hospital Roosevelt in Guatemala City.

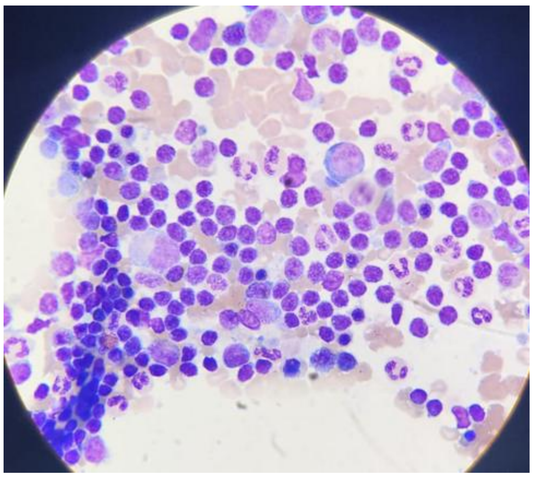


Photo courtesy of Dr. Leire Montoya

Results

The analyzed population consisted in 27 patients, 56% were female and 44% male. Average age of diagnosis was 68 years old (49-96). The average total lymphocyte count was 72 K/ul, average hemoglobin level 10.5g/dL and average platelet number of 253 K/ul. 56% of the patients had mutated IGHV, and 52% had at least 1 cytogenetic alteration. The most common mutation was del(13q), which was detected in 33% of the population, followed by trisomy 12 in 15%. TP53 mutation was detected in 2 patients.

Table 1. Patients Characteristics

No. of Patients	27
Age, Median (range), year	68 (49-96)
Sex N (%)	
Male	12 (44)
Female	15 (56)
Average of Total lymphocyte count	72 x10^9/L
Average Hemoglobin level	10.5g/dL
Average Platelet count	273 x10^9 L
Mutated IGHV (%)	15 (56%)
Most common Mutations (%)	14 (52%)
Del13p	9 (33%)
Trisomy 12	3 (12%)
TP 53	2 (7%)

Conclusions

Mutational status of IGHV and cytogenetic abnormalities in patients with CLL are important for risk stratification and prognosis. We have similar incidence of mutated IGHV, but have less cytogenetic abnormalities compared to what it is described in Western countries.

References

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