Genetic and Mutational Characterization in Patients with Chronic Lymphocytic Leukemia at the Guatemalan Social Security Institute

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Introduction

Chronic lymphocytic leukemia (CLL) is the most frequent leukemia in developed countries. Most Latin American countries do not have data of the epidemiology and mutational status of this pathology.

This is the first study of its kind in Guatemala to identify the most frequent mutations in a hospital in Guatemala and offer better treatment options.

Aim

- To establish an epidemiological baseline in the Guatemalan population, in order to improve therapeutic options, including iBTK and iBCL-2.

Objectives

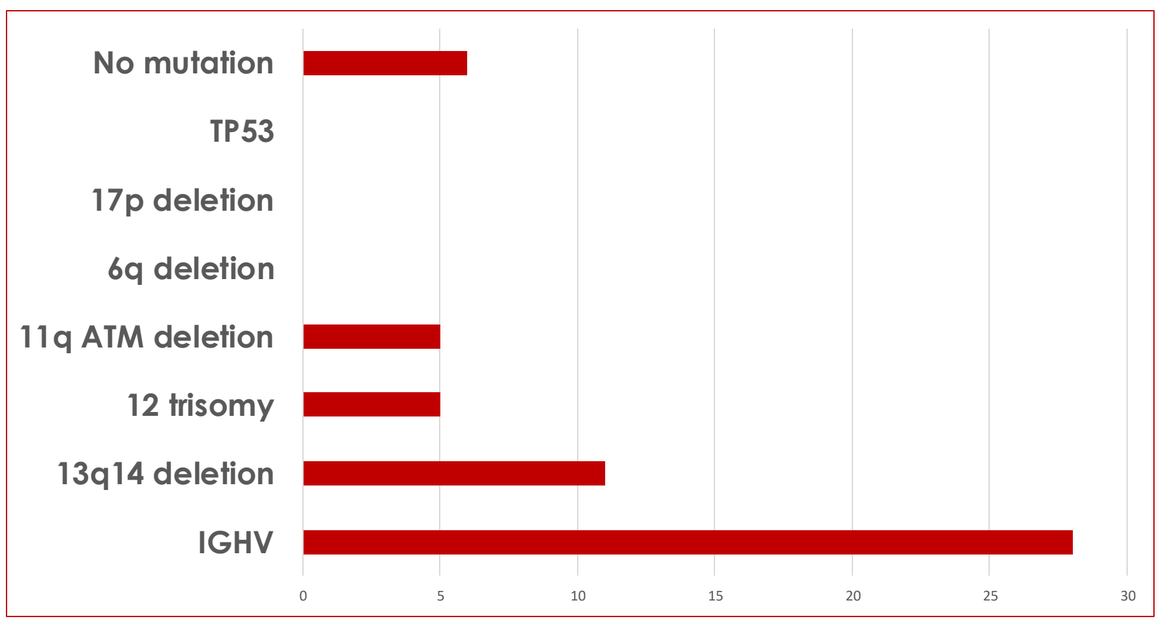
- Identify genetic mutations in patients with CLL and determine the incidence of CLL at Guatemalan Social Security Institute (IGSS).

Methods

- **Design**:
  observational, retrospective, cross-sectional, and analytical study.
- **Data collection**:
  between January 1st, 2020 and December 31st, 2023.
- **Participants**:
  Adults over 18 years of age with affiliation to IGSS.
- All patients were diagnosed by flow cytometry, then FISH and, lastly PCR for determination of IGHV somatic hypermutation and TP53 mutations.
- **Statistical analysis**:
  Chi-square was used for categorical variables with and $\alpha = 0.05$.

Results

- 40 patients were diagnosed between 2020 an 2023.
- Median age was **64 years**.
- Incidence for this population was **0.3/100,000** persons per year.
- A distribution of 1.2:1 ratio of females and males was found respectably.
- No **TP53** mutation, **17p** deletion or **6q** were found.
- Six patients with no mutations were identified.
- No association between mutational status and sex (p= 0.67) either patients age (p= 0.53) were found.



Discussion

Distribution of mutations is similar to that of the global population. The IGHV mutation is the most common.

Poor prognosis mutations such as TP53 and 6q deletion were not found, may be due to low incidence.

No associations were identified between mutational status and the clinical variables evaluated as in the rest of the world.

Conclusions

- Incidence was lower compared to the rest of the world, even other Latin American countries.
- Most patients had good prognostic mutations, no mutations with poor prognostic were found.
- No association between CLL and demographic characteristics were found.

References

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