Pediatric Acute Lymphoblastic Leukemia: incidence of complex karyotype by subgroup

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INTRODUCTION

Acute lymphoblastic leukemia (ALL) is characterized by the presence of chromosomal abnormalities in lymphoid cell lineage. Traditionally, this disease is classified according to immunophenotype into B or T cells, and subdivided into pre-B, pro-B and common. Cytogenetic alterations detected by karyotyping provides a better perspective on prognosis of disease (Wafa, A., et al). Current treatments provide favorable recovery expectation in more than 90% of cases, however the presence of complex karyotype is associated with poor outcomes.

AIM

The aim of the present study was to established the incidence of complex karyotype within ALL subtypes in a pediatric population and the associated cytogenetic abnormalities.

METHOD

The study includes a retrospective cross-sectional analysis of data from pediatric population (aged 3 months to 19 years) diagnosed with ALL during a three year period referred from health institutions within Mexico. Cases were classified according to immunophenotype and karyotype was obtained from cell cultures according to standard GTG methodology**.**

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CONTACT INFORMATION

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RESULTS

One hundred and eight cases of a pediatric population with ALL diagnosis were included in the study. Eighty-seven cases corresponded to the pre-B subtype (82%), the other subtypes, pro-B, T-ALL, and nonspecific accounted for 10, 7 and 1% respectively (**Figure. 1**). Thirty-five percent of the cases (38) had an abnormal karyotype with a single abnormality. Twenty-eight cases were classified as complex karyotype with three or more abnormalities, most of them (79%) included in the pre-B subgroup (**Figure. 2.A**). Chromosome loss and gain were the most frequent anomalies in this type of karyotype. Most frequent chromosomal alterations were +21 (6%), +X (6%) and +14 (5%) (**Figure. 2. B**).

CONCLUSIONS

Complex karyotypes were present in 79% of ALL patients classified as pre-B subtype. The predominant markers were +21, +X and +14 in the complex karyotypes. Interestingly, translocations were less frequent. Concerning to DNA index, most values correspond with the chromosome number in the karyotype, but there are some exceptions in pre-B cases (**Table I**). Integral diagnostics is recommended for a better understanding of each case.

REFERENCES

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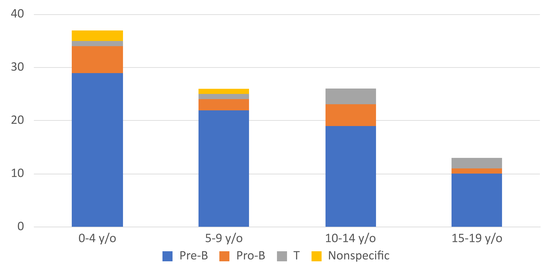


Figure. 1. Age and immunophenotype distribution of cases. The predominant presence of the Pre-B immunophenotype in all age ranges should be highlighted.

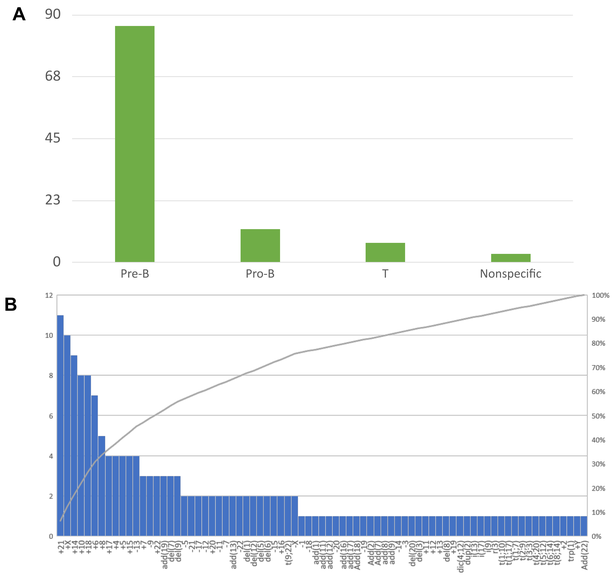
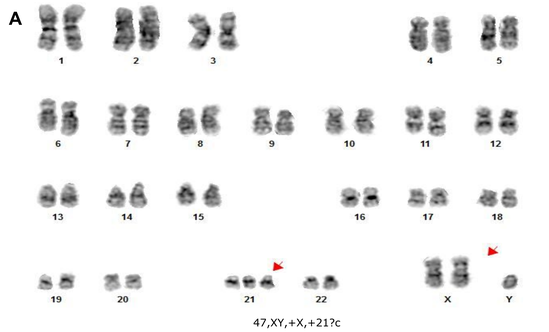


Figure. 2. Complex karyotypes distribution: all cases. A) Number of cases by immunophenotype. **B)** Chromosomal markers detected in complex karyotype (chromosome loss and gain were the most frequent anomalies).



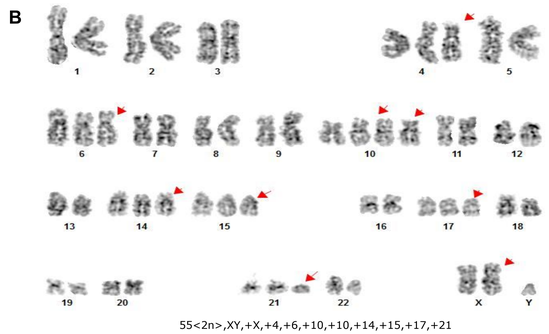


Figure. 3. Representative G-Band Karyotype A) Abnormal karyotype presenting gain +X and +21. **B)** Complex karyotype with multiple abnormalities (hyperdiploid)

Table I. Characterization by ploidy and DNA index of subgroup cases

Immunophenotype	Euploidy Hypodiploidy (≤46)	Hyperdiploidy (≥ 47)	DNA Index ≤ 1.14	DNA Index ≥ 1.15
Pre-B (n=23)	12	11	17	6
Pro-B (n=4)	3	1	3	1
T (n=1)	1	0	1	0