The interest of standard and molecular cytogenetics for the diagnosis of acute lymphoblastic leukemia in children

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Abstract

Introduction: Leukemia is a malignant proliferation of lymphoid cells blocked at an early stage of their differentiation that can invade the bone marrow, blood, and extramedullary sites. It is due to an underlying genetic alteration that affects many genes that encode proteins and play a crucial role in developing lymphoid cells. The study's objective is to determine the role of the standard karyotype and molecular biology for the diagnosis of ALL. **Patients and methods:** We conducted a retrospective study over 13 years, between January 2006 and December 2019, at the hemato-oncology unit at Abderrahim Harouchi's university children's Hospital in Casablanca. All the patients diagnosed with ALL de Novo during this period were included in this study. Their data were collected from the oncology unit's registry, and the medical information were extracted from the files. Statistical analysis was performed. Their results were discussed and compared to the literature data in the diagnosis part. However, patients who died before chemotherapy or were transferred to another facility were excluded from the outcome analysis. **Results** We conducted a retrospective study over 13 years, between January 2006 and December 2019, at the hemato-oncology unit at Abderrahim Harouchi's university children's Hospital in Casablanca. An unsuccessful karyotype was observed in 24.7%, whereas a successful karyotype was found in 75.3 % of our patients. In the latter normal karyotype was observed in 54%, and an abnormal one was retrieved in 46%. Numeral abnormalities were found in 48% of the cases (especially hyperdiploidy). Structural abnormalities were observed in 36% of the cases, and complex karyotype in 16% of the cases. The relapse risk among patients with unsuccessful standard karyotype after the first line of chemotherapy was higher than in the group with a successful one. **Discussion:** Compared to the literature, the findings contribute widely to the diagnosis of successful karyotype and help to adjust the risk group, adapt the treatment and improve the outcome in children with ALL. The unsuccessful standard cytogenetic was observed with a significantly higher risk of relapse and death in the statistical analysis in this group of patients. Those results suggest the use of molecular cytogenetics such as FISH, RT-PCR, and SKY to go beyond the limits imposed by the resolution of the banding and reveal cryptic anomalies essentially in unsuccessful standard cytogenetic cases to find out the underlying genetic abnormality that might refine the diagnosis and improve the prognosis in children with leukemia. **Conclusion**: Standard cytogenetics is useful for the diagnosis and needs to be completed by molecular cytogenetics to refine the diagnosis, especially in unsuccessful cultures.

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