## **SUMMARY**

Acute lymphoblastic leukemia is the most common malignant disease affecting children, accounting for approximately 30% of childhood cancers. Before the advent of effective chemotherapy, ALL usually was fatal. With further progress of chemotherapy, the incidence of children whom they inter in complete remission increased.

Further progress in the treatment of ALL depends on the development of new techniques for identification of certain factors that affect prognosis of the disease. Patients with unfavorable clinical and laboratory features were considered high risk patients and intensive therapeutic strategies should be considered.

One of the most recent and important prognostic factors in estimating the prognosis was the chromosomal study of ALL patients.

Familial aggregation of leukemia has been noticed. Also the risk of leukemia in sibling of children with leukemia has been estimated to be more than that of normal population.

This prospective trial was conducted to study an Egyptian ALL patients. A standard epidimiologic data on all patients and cytogenetic analysis in the form of karyotypic study was performed. Also our study included the examination of the healthy members of the family of the affected children.

Adequate examination was applied for each patient including complete history and physical examination, chest X ray, Abdominal sonography, complete blood count, bone marrow examination and karyotypic study.

Out of the 30 cases studied, successful karyotype was performed in 22 cases. These 22 cases were classified into two groups; group I (group of good prognosis) and group 2 (group of bad prognosis). The prognosis was evaluated according to the follow-up for one year after initiation of treatment. Group 1 (group of good prognosis) implied 11 cases, 10 of them were of normal karyotype and one case was hyperdiploid  $\geq$  50 chromosome. Group 2 (group of bad prognosis) implied 11 cases two of them were hyperdiploid (from 46-49 chromosomes), two cases were hypodiploid and the remaining were of normal diploid number. We recorded 6 cases with translocations which were considered to be the most common structural chromosomal abnormality. Only one case had deletion and it was in the short arm of chromosome 9. A strong relation was found between chromosomal abnormalities, especially translocations, and bad prognosis.

Normal chromosomal patterns were recorded in the all healthy members of the leukemia children's families.

## **CONCLUSION AND RECOMENDATION**

## Conclusion:

- (1) The presence of any chromosomal abnormality is an indicator mainly of bad prognosis in all patients.
- (2) Translocation is the most worse type of chromosomal abnormality as a landmark of prognosis. The specific form of translocation, the ph + ve ALL patient also had poor treatment outcome.
  - (3) Hypodiploidy is associated also with poor therapy outcome.
- (4) Hyperdiploidy (47 49) and pseudodiploidy cases have relatively poor prognosis.
- (5) Patients with apparantly normal chromosome have relatively good prognosis.
- (6) Hyperdiploid  $\geq 50$  is the only chromosomal abnormality, detected in our study, which has relatively good prognosis.
- (7) There is no chromosomal abnormalities in the healthy members of leukemic children's families.

## Recomendations:

- (1) Cytogenetic analysis should be done to every leukemic children at diagnosis and also at relapse (if it occured).
- (2) If any chromosomal abnormality was detected (especially translocation), intensive chemotherapy program and bone marrow transplantation should be considered as therapeutic options.
- (3) Cytogenetic analysis should be considered as one of the procedures done in routine follow up for ALL children together with the ordinary bone marrow examination and pripheral blood picture.
- (4) Further follow up to the healthy members of leukemic children's families is recomended. This follow up must be performed for a long period as these members may develop leukemia later on.
- (5) Concerning the technique of cytogenetic analysis, we recomend certain modification in the ordinary karyotypic study. This modification give us a better results. Four samples will taken from the patients and incubation with growth media is performed for different times. In bone marrow the first sample should be analysed directly, the second after 24 hours, the third after 48 hours and the fourth after 72 hours. In pripheral blood the first sample should be analysed after 24 hours, the second after 48 hours, the third after 72 hours and the fourth after 120 hours. Wheareas these four samples were practically difficult to be taken from an ill child (as we had to obtain four bone marrow samples).

We hope to use an advanced technique in cytogenetic analysis in ALL children, that is the high resolution banding technique. It is a well proven fact that this approach has provided a more accurate diagnosis of chromosomal defect in human. This technique will provide us with a less condensed chromosome. The advantage of the elongated chromosome is that they can visualise finer details with large number of bands.

(6) Cytogenetic analysis team should be in available in every hospital or centr dealing with leukemia.