

CYTOGENETICS

Comparative Genomic Hybridization study of Myelodysplastic syndrome

Year of Commencement 2004

Year of completion *ongoing*

Background : The myelodysplastic syndrome (MDS) is a clonal disorder of hematopoietic stem cells and occurs due to progressive sytopenia, defects in erythroid myeloril and megakaryocytic maturation. The MDs is a elderly disease and the high incidence was reported in above 60 years of age group. We had already carried out a cytogenetic study (1999-2004) in MD's patients and majority (55%) of patients were below 45 years age group. The chromosomal abnormalities including normal chromosomal and abnormalities were detected in 35% of patients. A high frequency of chromosome breakage was detected in Karyotypically normal MDs patients. In view of our findings the study extended to study genomic imbalances using molecular cytogenetic methods especially comparative genomic hybridization with the following objectives.

Detection of copy number variation in the genome

Identification of new oncogene sites in the chromosomal regions.

During one year period cytogenetic study was carried out in 18 suspected MDs patients. The chromosomal abnormalities monoassay 5,7 and 7 Q deletions detected 5 () patient remains patients Karyotype was normal. The patients' samples also collected in EDTA tubes for ANA extraction. The CGH methods under standardization and will be applied in patients.

Cytogenetic study of Acute lymphoblastic leukemia in young.

Objectives:

- To detect the chromosome abnormalities by conventional cytogenetic method (GTG banding)
- Molecular cytogenetic evaluation in cryptic and complex translocations by FISH and CGH study in all the patients.
- Correlation of cytogenetic abnormalities with immunophenotyping and other hematological parameters.

Cytogenetic study was carried out in 12 de novo ALL patients who were below the 15 years of age. All clinical details were recorded in our Proforma. Chromosome preparation from bone marrow was done by direct method. The culture also set up with B and T specific lymphocyte stimulants on Blood and bone marrow samples. Chromosome analysis was done on GTG banded metaphases. Among these 12 patients 9 (75%) were successfully karyotyped. 7 (58%) patients had chromosomal FISH using 12; 21-probe will be applied in patients with normal karyotype with G-banding. The samples also collected in EDTA tubes for DNA extraction and CGH will be done on ANA samples.

Diagnosis	No. of patients	Type of chromosome abnormalities	No.
T - ALL	5 (41.7%)	Hyperdiploidy (>50 chromosomes)	2
		47,XY, + mar	1
		46,XY, del (9)(p22 -ter)	1
B - ALL	7 (58.3%)	46,XY, dup(1)	1
		Hypodiploidy	1
		46,XY/del(9)(p22-ter)/hypodiploidy	1
Total	12		7(58 %)