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Cytogenetic investigations in bone marrow cancers and cancer predisposing chromosomal instability syndromes

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Background: Leukemia is a neoplastic proliferation of hematopoietic cells. Leukemia can be classified as either myelocytic (myeloid) or lymphocytic (lymphoid) depending on the lineage of the leukemic cells and chronic and acute depending on the clinical course.

Aim: The aim of the present study was to detect structural abnormalities by using automated karyotyping (IKAROS) software in Leukemia cancer (CML, AML, ALL and CLL) samples.

Methods: Conventional cytogenetics: Peripheral Blood Culture (PBC) and bone marrow aspirate supplemented with mitogen Phytohemagglutinin (PHA) metaphase chromosomes were harvested after 72 hours for chromosome analysis. Image acquisition and analysis was performed by using automated karyotyping (IKAROS) software based on GTG banding.

Results: Philadelphia chromosome involving the reciprocal translocation between the long arms of chromosomes 9 and 22 (46, XY, t (9; 22)) was observed in majority of Chronic Myeloid Leukemia (CML) patients of the present study. Also a high number of chromosomal structural aberrations were observed in Fanconi Anemia (FA) patient, a chromosomal instability syndrome predisposing to cancer.

Conclusion: Conventional cytogenetics can be the method of choice in diagnosis of genetic conditions like CML but for other bone marrow cancers more studies with high sample numbers need to be carried out for determination of clear cut markers.

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