Use of chromosome painting and quantitative analysis of the fluorescent signal to detect small structural aberrations

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Advances in protocols for FISH and digital imaging microscopy made easier the detection of chromosomal rearrangements in metaphase chromosomes. In this work we present an easy and reproducible procedure that combines a protocol of chromosome painting modified by us and a relative quantitative analysis of the fluorescent signal on confocal microscope. We studied a patient with acute myeloblastic leukemia (AML) who presented in bone marrow some malignant cells with trisomy 21. We applied chromosome 21-specific DNA probe for painting. Unexpected findings were revealed which had not been detected by the conventional cytogenetic methods. Such findings were as follows: the painting alone suggests an amplification of the signal in two of the three chromosomes 21, which was confirmed by the relative quantitative analysis by software COSMOS (BioRed) of confocal microscope MRC-600. Our data offer evidence that amplification is a result of a 21 chromosomal abnormality, probably gene(s) amplification. The aim of this work is to discuss the utility of the application of the combination of these two techniques in clinical cytogenetics to detect small structural chromosomal aberrations in malignant hematological diseases.