

MOLECULAR CHARACTERIZATION OF ACUTE LEUKEMIA  
TRANSLOCATIONS. Hors CP, Caputo L, Mantesso A, Andrade V, Garicochea B.  
Laboratório de Genética Molecular do Hospital Sírio Libanês [genetica@sbsl.br](mailto:genetica@sbsl.br) ,  
[corapereira@hotmail.com](mailto:corapereira@hotmail.com)

Conventional metaphase analysis has been considered to be the “gold standard” for determining diagnosis of leukemia, but this technique normally requires bone marrow aspiration and is therefore invasive. The frequency of cytogenetic analyses can be considerably reduced if patients are also monitored by molecular methods, which can be performed on peripheral blood specimens. Of the various techniques available, most attention has been paid to RT-PCR for the analyses of unique breakpoint fusion regions in the transcript of the implicated translocated genes. The advantages of this method over conventional cytogenetics are its sensibility and sensitivity. A lack of culture and cellular replication induction is another technical advantage. Besides, this technique allows precise monitoring of minimal residual disease (MRD). With the aim of disponibilizing this methodology to improve the clinical decision-making in leukemia patients, we have tested leukemia carrying translocations cell lines both by cytogenetics and RT-PCR, to be utilized as positive controls in the molecular characterization of acute leukemia commonly occurring translocations. Using cytogenetics the t(1;19), t(4;11), t(8;21), t(9;22), t(15;17) and inv(16) can be accurately detected when metaphase is obtained. By RT-PCR all of them and also t(12;21), not visible under conventional cytogenetics, fusion gene transcripts are detected, besides del(1p32). We standardized RNA extraction and RT-PCR protocol to be used for molecular classification of acute leukemia at diagnosis and for MRD detection during follow-up to evaluate treatment effectiveness. It is envisaged that efforts in characterization of molecular defects in leukemia complementing conventional metaphase analysis or by itself in some cases, will ultimately be translated in better clinical outcome for patients. Órgão Financiador : não há