

Identification of chromosomal translocations in leukaemia using multiplex reverse transcriptase polymerase chain reaction: a retrospective ten years study in Malaysia

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Abstract

Leukaemia is the sixth most common cancer in Malaysian population and seventh most common in males and eighth most common in females. It is the most common cancer among children. Recent advances in genomics have contributed significantly towards a better understanding of the genetic landscape of leukaemia. Identification of recurrent chromosomal translocations which can be detected in a substantial number of these patients are important for classification of the disease, prognostication, treatment monitoring and also to guide targeted therapy. A total of 3433 bone marrow or peripheral blood samples were collected from patients who were newly diagnosed with leukaemia. Their demographic data, bone marrow morphology and also immunophenotyping results were recorded. Multiplex reverse-transcriptase polymerase chain reaction was performed using the HemaVision[®]-28N protocols for detection of 28 common translocations. This study aims to report the incidence of leukaemia-specific translocations in Malaysian patients who were admitted in tertiary care hospital from 2008 to 2017. Among the 3433 patients, 1500 patients were diagnosed with Acute Lymphoblastic Leukaemia, 1160 for Acute Myeloid Leukaemia, 581 for Chronic Myeloid Leukaemia and 192 patients for Acute Promyelocytic Leukaemia. We found that 34.7% of these patients have chromosomal translocations. Overall, 23 fusion gene transcripts were detected. The most common genetic aberration found were BCR-ABL1 47.8%, PML-RARA 13.1% and RUNX1-RUNX1T1 11.4%. Interestingly, based on our study, we found one CML case with unique breakpoint with larger PCR product compared to common breakpoint within BCR gene at exon e14 (B3) and ABL1 gene at exon e2 (A2). In this case, the PCR product have additional 124bp and sequencing confirmed the extra nucleotide derived from ABL1 gene at exon 1 chromosome 9. Multiplex RT-PCR is an effective and rapid screening tool for detection of recurrent chromosomal translocations in leukaemia. A comprehensive sub classification of leukaemia by molecular technique is very useful not only for diagnostic purpose, but also for risk stratification, prognostication and targeted therapy.

Keywords: Leukaemia, chromosomal translocation

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