JKMS

http://dx.doi.org/10.3346/jkms.2011.26.10.1401 • *J Korean Med Sci 2011; 26: 1401*

The Author Response

Diagnostic Standardization of Leukemia Fusion Gene Detection System using Multiplex Reverse Transcriptase-polymerase Chain Reaction in Korea

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We appreciated Dr Park TS for his interest and comments in our recent paper (1) in *Journal of Korean Medical Science* regarding a diagnostic standardization and detection methods of leukemia-associated gene rearrangements in Korea. Our study demonstrated the spectrum and frequency of chromosomal abnormalities in patients with mainly acute leukemia, which were differed from previous studies. Also, this study may offer important implications in the de-velopment of new molecular detection system for screening panel, as well as revisions of the current commercially available multiplex RT-PCR system. New molecular detection system for screening panel in our study means variety of molecular biological methods including PCR-based system, fluorescence in situ hybridization (FISH) and others. We agree that the FISH assay is better suited rather than the RT-PCR method in t(3;3)(q21;q26), inv(3)(q21q26), t(8;14)(q24;q32), and i(17)(q10).

However, one can use RT-PCR detection system for screening the aforementioned chromosomal abnormalities based on the following backgrounds. First, the protocol of RT-PCR assay for the detection of either inv(3)(q21q26) or t(3;3)(q21;q26) was already published (2). This protocol might be used in multiplex RT-PCR system for the detection of leukemia fusion genes. Second, it is well known that the t(8;14)(q24;q32) (c-*myc*/IgH rearrangement) has no fusion transcript and fusion protein (3). So, the c-*myc*/IgH rearrangement is only detectable at the DNA level (the RNA expression of c-*myc* (exon 1 or exon 2) and/or IgH (constant and joining region) also might be absent or remarkable depressed). Based on the fact of decrease or absence of RNA expression of these genes in the case of t(8;14)(q24;q32), conventional RT-PCR and quantitative RT-PCR for them might be implemented for screening. Genomic DNA PCR (long-distance) assay for the detection of the t(8;14)(q24;q32) was already established using one primer for the c-*myc* gene in exon 2 and four primers for the IgH locus (3, 4). Third, similarly, the screening of i(17)(q10) chromosomal abnormality by semiquantitative RT- PCR assay might be possible based on the absence of expression of p53 gene located at 17p13.1 due to deletion of short arm of 17 chromosome.

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pISSN 1011-8934
eISSN 1598-6357