CASE 038

CŌRE DIAGNOSTICS™

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A 25 year old female who was clinically diagnosed as CML was referred to our cytogenetic laboratory for chromosomal analysis.

MATERIAL AND METHOD

Chromosomal analysis was performed on 2 ml heparinized Bone marrow sample by 24 and 48 hours unstimulated cultures with appropriate serum, antibiotics and without Phytohemagglutinin[1]. A total of 20 metaphases were analyzed through GTG-banding (G bands by trypsin and Giemsa) according to International System for Human Cytogenetic nomenclature2016 [2,3]. All Images were captured using fully automatic Olympus BX63 microscope and analyzed using Bio-view karyotyping software. The FISH analysis was also performed according the standard protocol from sample for confirmation of BCR-ABL gene using dual color fusion FISH probe (Cytotest probe). All cells images were captured using Olympus fluorescent microscope BX-61 equipped with a CCD camera and analyzed using Bio-view FISH software.

FINAL DIAGNOSIS

The results showed balanced translocation between the short arm of chromosome # 1 and short arm of chromosome # 4, another translocation involving three chromosome at bands short arm of chromosome # 6, long arm of chromosome # 9 and long arm of chromosome # 22. The karyotype of the patient was 46,XX,t(1;4)(p32;p16),t(6;9;22)(p23;q34; q11.2)[20] (Figure 1). BCR-ABL fusion also confirmed by FISH testing and result was POSITIVE as 1 Fusion 2 Green and 2 Orange (Complex pattern) (Figure 2).