CASE 015

CŌRE DIAGNOSTICS™

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CLINICAL HISTORY

A 5-months-old male child was referred to our cytogenetic laboratory for chromosomal analysis. He was the second baby of non-consanguineous parents. Family history was not significant. On physical examination, dysmorphic facial features, including mongoloid facies, cleft palate, simian crease, and developmental delay were observed.

PATHOLOGIC FINDINGS: Chromosomal analysis was performed on 2 ml heparinized peripheral blood sample by 72 hours stimulated cultures with appropriate serum, Phytohemagglutinin, and antibiotics[1]. A total 30 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 Olympus BX51 microscope and analyzed using Bio-view karyotyping software. The results showed balanced translocation between short arm of chromosome # 3 and short arm of chromosome # 10. The karyotype of the child was 46,XY,t(3;10)(p21; p13) (Figure 1).

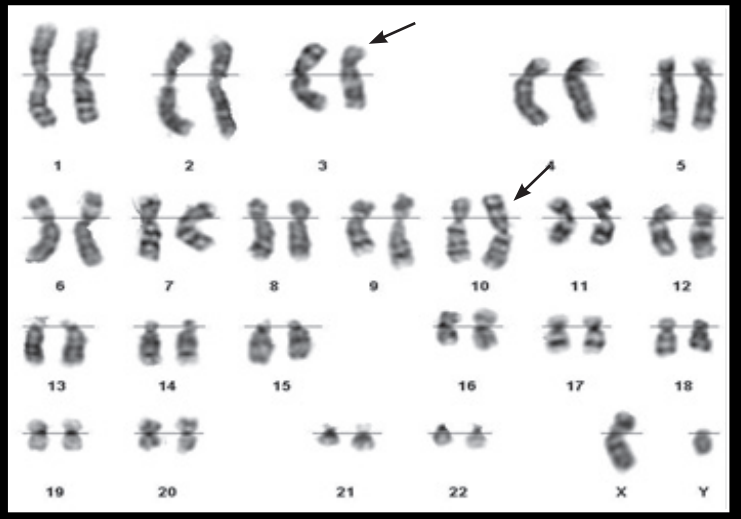

Figure 1