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A Report of 5-Months-Old Baby with Balanced Chromosomal Rearrangements along with Phenotypic Abnormalities

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Abstract : We report here a case of five-months old male baby, born as second child of non-consanguineous parents with no considerable history of genetic abnormality which was referred to our cytogenetic laboratory for chromosomal analysis. Physical dysmorphic facial features including mongoloid face, cleft palate, simian crease, and developmental delay were observed. We present this case with unique balanced autosomal translocation of t(3;10)(p21;p13). The risk of phenotypic abnormalities based on de novo balanced translocation was estimated to be 7%. The association of balanced chromosomal rearrangement with Down syndrome features such as multiple congenital anomalies, facial dysmorphism and congenital heart anomalies are very rare in a 5-months old male child. Trisomy-21 is not uncommon in chromosomal abnormality with the birth defect and balanced translocations are frequently observed in patients with secondary infertility or recurrent spontaneous abortion (RSA). Two ml heparinized peripheral blood cells cultured in RPMI-1640 for 72 hours supplemented with 20% fetal bovine serum, phytohemagglutinin (PHA), and antibiotics were used for chromosomal analysis. A total 30 metaphases images were captured using Olympus-BX51 microscope and analyzed using Bio-view karyotyping software through GTG-banding (G bands by trypsin and Giemsa) according to International System for Human Cytogenetic Nomenclature 2016. The results showed balanced translocation between short arm of chromosome # 3 and short arm of chromosome # 10. The karyotype of the child was found to be 46,XY,t(3;10)(p21; p13). Chromosomal abnormalities are one of the major causes of birth defect in new born babies. Also, balanced translocations are frequently observed in patients with secondary infertility or recurrent spontaneous abortion. The index case presented with dysmorphic facial features and had a balanced translocation 46,XY,t(3;10)(p21;p13). This translocation with break points at (p21; p13) has not been reported in the literature in a child with facial dysmorphism. To the best of our knowledge, this is the first report of novel balanced translocation t(3;10) with break points in a child with dysmorphic features. We found balanced chromosomal translocation instead of any trisomy or unbalanced aberrations along with some phenotypic abnormalities. Therefore, we suggest that such novel balanced translocation with abnormal phenotype should be reported in order to enable the pathologist, pediatrician, and gynecologist to have a better insight into the intricacies of chromosomal abnormalities and their associated phenotypic features. We hypothesized that dysmorphic features as seen in this case may be the result of change in the pattern of genes located at the breakpoint area in balanced translocations or may be due to deletion or mutation of genes located on the p-arm of chromosome # 3 and p-arm of chromosome # 10.

Keywords : balanced translocation, karyotyping, phenotypic abnormalities, facial dimorphisms
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