

In the index case, we observed balanced chromosomal translocation instead of trisomy or unbalanced aberrations along with some phenotypic abnormalities. Therefore, we suggest that any such novel balanced translocation with abnormal phenotype should be reported in order to enable the pathologist, geneticist, pediatrician, and gynecologist to have a better insight into the intricacies of chromosomal abnormalities and the associated phenotypic outcomes/features. It is hypothesized that the 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.

REFERENCES

- 1. Kumar M, Thatai A, Chapadgonkar S. Homozygosity and Heterozygosity of the Pericentric Inversion of Chromosome 9 and its Clinical Impact. Journal of Clinical and Diagnostic Research 2012;6:816-820.
- 2. Seabright, M. A rapid banding technique for human chromosomes. Lancet 1971;2;971.
- 3. L.G. Shaffer JM-J, M. Schmid. ISCN An International System for Human Cytogenetic Nomenclature. S. Karger, Basel 2016.
- 4. Akgul M, Ozkinay F, Ercal D, Cogulu O, Dogan O, Altay B, Tavmergen E, Gunduz C, Ozkinay C. Cytogenetic abnormalities in 179 cases with male infertility in Western Region of Turkey: report and review. J Assist Reprod Genet. 2009;26:119–22.
- 5. Warburton D. De novo balanced chromosome rearrangements and extra marker chromosomes identified at prenatal diagnosis: clinical significance and distribution of breakpoints. Am J Hum Genet. 1991;49(5):995-1013.
- 6. Reardon W, Donnai D. Dysmorphology demystified. Arch Dis Child Fetal Neonatal Ed 2007;92:225-229.
- 7. Haldeman-Englert CR, Saitta SC, et al. Evaluation of the dysmorphic infant. In: Gleason CA, Devaskar SU (9th ed) Avery's diseases of the newborn. Philadelphia: Elsevier Saunders2012;186-195.
- 8. Cotter PD, Caggana M, Willner JP, Babu A, Desnick RJ. Prenatal diagnosis of a fetus with two balanced de novo chromosome rearrangements.

 Am J Med Genet 1996; 66: 197-9.