

Inherited Unbalanced Chromosome from Parent with Balanced Translocation: A Case Report and Review of Literature

Kumari P^{1*}, Mishra VV¹ and Tewari S²

Abstract

Reciprocal translocations are common, and the translocation heterozygote (carrier) may have a risk to have a child who would be mentally and physically abnormal due to a segmental aneusomy. The carriers of balanced translocation mostly do not have recognizable phenotypic expression, however, they can produce unbalanced chromosome, which is transmitted to next generation through fertilization of gametes carrying the derivative chromosome. We describe a 4-year-old boy with partial 10q trisomy and distal 13qmonosomy. The patient presented with delayed milestones, dysmorphic face, congenital heart defect, renal and skeletal anomalies. The conventional cytogenetic analysis showed a 46,XY, add(13q) karyotype. The child inherited the unbalanced chromosome from mother who was a carrier of balanced reciprocal translocation,t(10;13)(p24.2;q33.1). The phenotype observed in our patient resulted from the combination of those defects described in the isolated dup(10q) and distal del(13q) syndromes but predominantly resembles the children with distal trisomy 10q syndrome. Our finding are in concordance with the literature and supports the importance of critical regions like 10q24 and 13q32 for the phenotypic expression of distal 10q trisomy syndrome and distal 13q trisomy syndrome respectively.

Keywords

Balanced reciprocal translocation; Distal trisomy 10q; Distal trisomy 13q