

Tetrasomy X in a Child with Upper Limb Deformity

49, XXXXY syndrome is a rare sex chromosome aneuploidy in humans with an incidence of 1:85000 male births [1]. Various clinical features such as low birth weight, slow growth with reduced bone age, mental retardation, radioulnar synostosis, multiple skeletal deformities with joint laxity, clinodactyly, cardiac deformity, abnormal genitals, hypogonadotropic hypogonadism, and craniofacial anomalies have been reported previously [2,3]. A 16-months-old male child, born to non-consanguineous parents, was referred to our laboratory for chromosomal analysis. The mother's age was 22 years and father was 35-years-old. The child had dysmorphic features (hypertelorism, upward slanting eyes, broad nose, low set small ears with tags, flat occiput), hypoplastic nipples, bilateral clinodactyly, cubitus varus (deformity in both elbows), flat feet, delayed dentition, and global developmental delay. Ultrasonography (USG) of abdomen revealed enlarged spleen and both the kidneys were normal. 2-D echocardiogram was normal. Cytogenetic analysis of G-banded metaphases from PHA-stimulated peripheral blood revealed 49, XXXXY karyotype (**Fig. 1**). Fluorescence in situ hybridization (FISH) using centromeric probes of X and Y confirmed tetrasomy X, and there was no indication of mosaicism.

Tetrasomy X occurs due to two non-disjunction events in maternal meiosis. Spectrum of clinical features have been reported in 49, XXXXY syndrome [2-4]. The clinical diagnosis of tetrasomy X in newborns is challenging. Chromosomal analysis plays an important role in accurate diagnosis. The phenotypic presentation in elderly patients with tetrasomy X is not known. Follow-up studies are essential in such rare cases to establish genotype-phenotype correlation. However, the additional



FIG.1. Metaphase showing tetrasomy X and Y.

X chromosomes and presence of Y chromosome may influence the development of Klinefelter syndrome phenotype in tetrasomy X syndrome. Genetic counselling for the parents is essential for the future pregnancies.

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