Case Report

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Partial Trisomy of 13q and Partial Monosomy of 6q; A Patau Syndrome Variant

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Abstract

Trisomy 13 often presents with classical features of Patau Syndrome. Atypical features are likely to suggest a mixed phenotype; like associated 6q monosomy. In this case, skeletal features associated with 6q monosomy domnated the phenotype.

Keywords: Patau Syndrome; Phenotype.

Introduction

Trisomy 13, also known as Patau syndrome, is a relatively common chromosomal condition in which there are three copies instead of the usual two copies of all, or a part of chromosome 13 in the cells of the body [1]. But the partial trisomy 13q is uncommon with few cases being described with a specific phenotype with extensive variability of expression [2]. The 6q terminal deletion syndrome is characterized by specific craniofacial dysmorphisms, short neck, and neurologic manifestations, along with various nonspecific malformations [3]. Association of 13q trisomy with 6q deletion with mixed phenotype is extremely rare with only one case by Fryns et al in 1974 [4]. Here, we present this rare entity of Partial trisomy of 13q and partial monosomy of 6q in a child, second of its kind.

Case Report

A four month old boy, 3rd by birth order, born of non consanguineous marriage, presented to outpatient department with complaint of cough, runny nose for two days and not gaining weight. He had normal amtenatal and perinatal history, being a full term normal vaginal delivery with birth weight 3 kg. At 4 month age child had social smile and partial neck holding. There was no significant family history and child had two normal elder sisters without any congenital abnormality.

On examination child had lethargy with central cyanosis but no clubbing. At 4 month of age, child weighed 3.3kg and length was 45cm; both below 3rd centile for age. Trigonocephaly was strikingly obvious with left occipital plagiocephaly and craniosynostosis of coronal suture and metopic suture. Anterior fontanel was barely open and head circumference was 39cm. Bushy eyebrows, hypotelorism, prominent nasal bridge, long philtrum, thin upper lip, micrognathia, malformed ear with short neck, ankyloglossia, short fourth metatarsal with right lumbar bony prominence were additional dysmorphic features (Figure 1a). Pupils and fundus were normal. There was central and axillary hypotonia.

Systemic examination was not contributory; there was no murmur, nor any focal neurodeficit or limb deformities or spina bifida. Opitz C-trigonocephaly syndrome, carpenter syndrome were thought as close phenotypical differentials. The results of the complete blood count tests, liver and renal function tests were normal. The patient had boot-shaped appearance of heart on the chest radiograph and L4 hemivertebra

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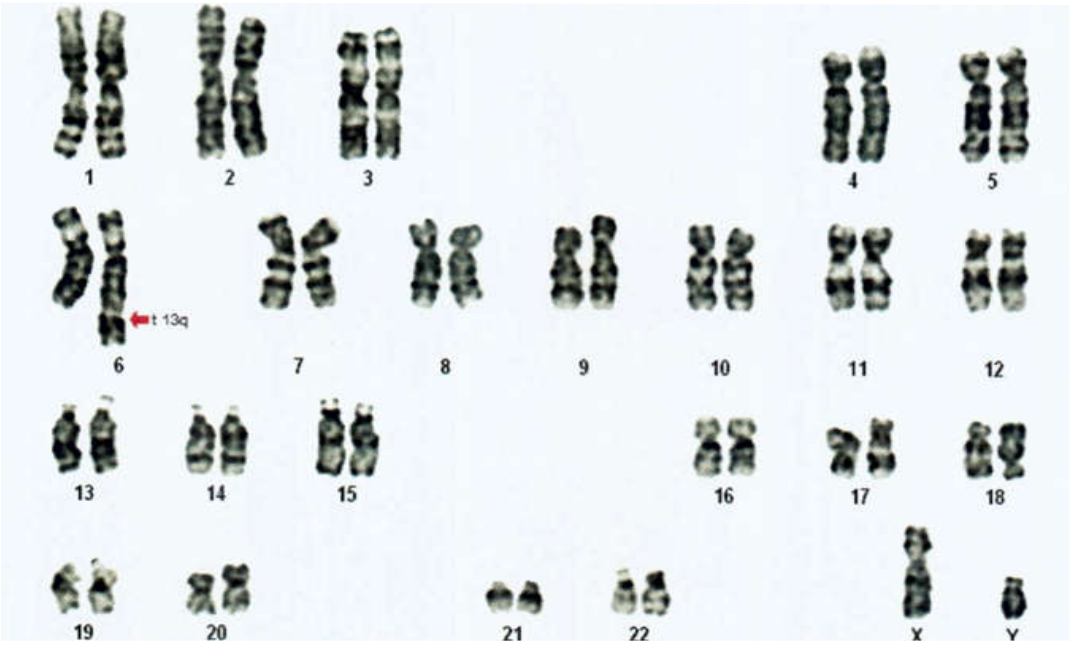
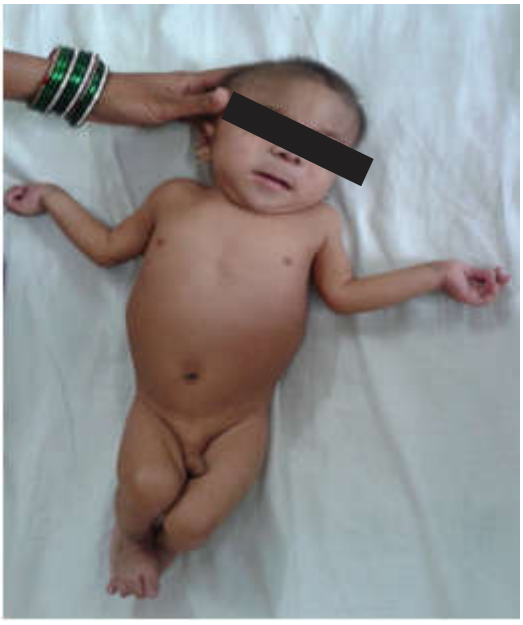
on X-ray abdomen (Figure 1b). The echocardiography revealed Tetralogy of Fallot's with severe pulmonary stenosis with mild pulmonary hypertension. The cranial computerized tomography revealed metopic synostosis (trigonocephaly) with prominence of left lateral ventricle, temporal horns and 4th ventricle. Chromosome analysis demonstrated caryotype of 23 XY, with gain of 13q 14.11-q ter segment and loss of 6q 22.1-q ter segment (Partial trisomy of 13q and partial monosomy of 6q) from peripheral blood leukocytes using 'G' banding technique (Figure 1c). Symptomatic treatment given for heart disease and upper respiratory tract infection and then the patient referred for surgical management.



Leucocyte Culture Metaphase Karyotype-G Banding



Karyotype depicting monosomy 6q and trsomy 13 and clinical photograph of the case

Discussion

Com-mon phenotypic features with terminal deletions involving chromosome 6q include intellectual disability, hypotonia, epilepsy, cardiac defects, retinal abnormalities, ear anomalies, facial dysmorphisms, and malformations of the brain, spinal cord, and vertebrae. Structural brain malformations are consistently observed in these patients and include agenesis of the corpus callosum (ACC), periventricular nodular heterotopia (PNH), polymicrogyria, hydrocephalus, and cerebellar [5]. Microcephaly is also a common feature. Facial dysmorphisms is described in form of large and malformed ear, micrognathia, high arched palate, long philtrum and short neck [6].

Our patient shared many of these craniofacial anomalies including metopic synostosis (trigonocephaly). Significant brain malformation with prominence of left lateral ventricle, temporal horns and 4th ventricle was present. Also, vertebral anomaly and congenital heart defect was present as he had L4 hemivertebra and Tetralogy of Fallot.

Partial trisomy 13q may result from parental reciprocal translocations; parental pericentric inversions or de novo direct duplications [7]. Partial trisomy 13q has been shown to have both a distinctive

and common pheno-type resembling that of complete trisomy 13. Also, there are distinctive clinical features between the trisomy of the proximal and distal regions of the long arm of chromosome 13[8]. Com-mon phenotypic features of partial trisomy 13q are: craniofacial dysmorphism (bushy eyebrows, long curled eyelashes, prominent nasal bridge, long philtrum, thin upper lip, microceph-aly, and hypotelorism), high arched palate, short neck, haemangioma, hexadactyly, urinary tract or kidney anomalies, umbilical or inguinal hernia, intra-uterine growth retardation, and oligohydramni-os. Other phenotypic features in child and adult patients described are: psychomotor retardation, hypoacusia, hypochromic anaemia, splenomegaly, ocular anomalies, convulsions, and fatty acid disturbances[7]. Our case despite having a karyotype of partial trisomy13 did not show most features of Patau syndrome or it's variant.

This case had presentation more similar to partial 6q deletion. Absence of most of phenotype of trisomy 13 insisted us to call it as a variant of Patau syndrome, probably a segmental translocation of 13q with segmental deletion of 6q. A similar case was published in 1974 by Fryns et al had features consistent with 6q deletion syndrome [4]. That makes our case a second of its type.

Conclusion

Cytogenetic abnormalities do decide phenotype. In case of mixed defects, they may not be consistent to any specific known syndrome. A partial trisomy 13, devoid of classical features of Patau syndrome is still a possibility; primarily due to deleted segment from other chromosome. So the features of Patau syndrome in a translocation need not be due to trisomy but due to primary deleted segment phenotype.

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