Case report

Edward's syndrome with a novel karyotype

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Abstract

Edward's syndrome was first described as a clinical entity in 1960 as a disorder of trisomy 18 (47 XX/XY; + 18) in babies with particular pattern of malformations. The Karyotype found in our case was (47 XX + 18 add (22) (p13) which has not been published so far in the literature. The less common findings noted in the baby were rocker bottom feet, syndactyly of 2nd and 3rd toes, microcephaly and corneal opacities. Though we didn't find any significant association between phenotypic ranges with genotypic variation in literature, but further research is needed for it. We are reporting this case as the genotype is found to be novel.

Keywords: Edward's syndrome, karyotype, neonate.

Introduction

Edward's syndrome was first described as a clinical entity in 1960 as a disorder of trisomy 18 (47 XX/XY; + 18) in babies with particular pattern of malformations.[1] Children with this syndrome commonly have intrauterine growth retardation (IUGR), feeding difficulty, flexion of the fingers, overlapping the index and the 3rd finger, cryptorchidism, prominent occiput, low set malformed ears and micrognathia. Less common features are epicanthal folds, small feet, rocker bottom foot, soft tissue syndactyly of 2nd and 3rd toes, microcephaly, corneal opacity, ptosis, cleft lip and cleft palate.[2] It is also associated with major organ anomalies like congenital heart disease, omphalocele and horseshoe kidney. Incidence of Edward's syndrome is 1 in 3000 to 7000 births and survival beyond infancy is unusual. There is a female preponderance with a ratio of 3:1[1]. Various karyotype patterns have been reported in literature, we are reporting a novel karyotype pattern not published till now.

Case report

Our case was a term, female baby with severe IUGR with ponderal index of 1.6 and birth weight 1200 grams. The baby was delivered vaginally at home. The maternal age was 28 years. She was 3rd gravida with one living healthy male child and previous history of one spontaneous abortion. The baby had overriding fingers, rocker bottom feet, second and third toes soft tissue syndactyly, microstomia, micrognathia, microcephaly & small feet. Ocular examination revealed bilateral corneal opacities. Baby had severe respiratory distress since birth due to meconium aspiration. CNS examination revealed hypotonia. Neonatal reflexes were sluggish.

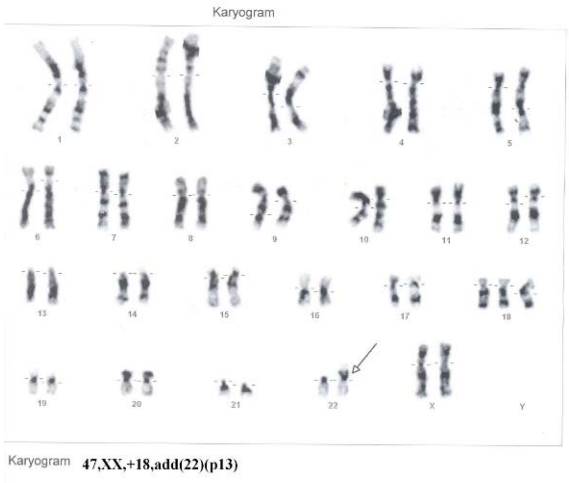

Figure 1

There was no apparent cardiovascular abnormality, respiratory examination was normal and abdominal examination did not reveal any organomegaly. Chest x-ray showed infiltrates in bilateral lung fields with no cardiomegaly. Abdominal ultrasound didn't reveal any renal anomalies. Echocardiography couldn't be done as the baby

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expired on second day of her life. Karyotype revealed presence of trisomy 18 with addition of extra chromosomal material of unknown origin at band p13 on chromosome 22 (47 XX + 18 add (22) (p13) (Figure 1).

Discussion

Edward's syndrome occurs due to fetal trisomy 18 and majorities are complete trisomy. It may be due to a nondisjunction as seen in elderly maternal age groups or translocations as seen in young mothers.[3] A common karyotype associated with Edward's syndrome is 47, XX, + 18 i(18q) & is associated with better prognosis. Partial trisomy of chromosome 18 like 18q2, 18p & q1 also has been reported. The phenotype ranges from mild presentation with no internal organ malformation to the classic presentation of this syndrome.[4] There are few cases reported with partial trisomy 18q due to maternal reciprocal translocation.[5] One case was diagnosed prenatally who had trisomy 18 along with monosomy of X chromosome.[6] In addition, there have been cases with double trisomy like 48, XYY, + 18 or 48, XXX, + 18.[7] Trisomy 18 has also been reported with pseudodicentric isochromosome formation like 46, XY, I dic(18)(p11::p11) 11or 47, XX, -18,+ i(18q) + i pseudic (18p) where both p & q arms had isochromosome[8].

We feel this is a unique case of Edward's syndrome with a novel Karyotype (47 XX + 18 add (22) (p13) having phenotypic features of rocker bottom feet, syndactyly of 2nd and 3rd toes, microcephaly and corneal opacities. This, however, needs further confirmation following in depth review.

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