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Case Report

A rare case report of unusual presentation of Edward's syndrome (trisomy 18)

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ABSTRACT

Edwards syndrome, a rare genetic disorder is characterized by the extra copy of chromosome 18. About 50% babies with this syndrome do not survive one week of age and approx. 95% do not survive past the first year of life. The syndrome is usually characterized by dysmorphic facies, microcephaly, flexion finger deformity and rocker- bottom feet. There is involvement of cardiacvascular and renal system with intellectual disability. Authors report a case of Edwards syndrome presenting with failure to thrive and developmental delay in the absence of usual clinical features of Edwards syndrome.

Keywords: Flexion finger deformity, Intellectual disability, Rocker-bottom feet, Trisomy 18

INTRODUCTION

Trisomy 18, also known as Edwards syndrome, is defined as an autosomal hereditary disorder, presents with an extra chromosome 18 in the karyotype study. It is the second most common autosomal disorder among live-born children after Trisomy 21. The incidence is one in every 6000 live births.[1] Of these, only 5% children live more than 1 year.[2] There is a 3:1 ratio in the prevalence of females to males, as most male infants with this disease die during pregnancy.[3] This syndrome affects multiple organs, leading to neurological, cardiac, pulmonary, gastrointestinal, musculoskeletal manifestations. This case is reported due to its unusual presentation and rarity of syndrome in Indian scenario.

CASE REPORT

A two year male child presented in the paediatric department with the complaints of inability to sit and stand, and not growing well. He was the only child born to non-consanguineous marriage. There was a history of abortions in previous pregnancies. His father and mother were 25 and 23 years old respectively at his birth. He was born at full-term by a vaginal delivery at institute with a birth weight of approx. 1850 grams.

The antenatal period was uneventful. The parents give the history of delayed cry for which the child was admitted in special new-born care unit for seven days and was given oxygen supplementation. The child was developmentally delayed. At the time of presentation, the child weighed 7.2kgs, height of 74 cm and head circumference of 47.5 cm.

On general examination, prominent forehead, low set ear, a small jaw, high arched palate, malocclusion of teeth, an upturned nose, unilateral ptosis, depressed nasal bridge was present (Figure 1).

Ear examination showed stenosis of external auditory canal. There was absence of palmar flexion creases (Figure 2). Laxity of both wrist joint and metacarpophalangeal joint of all fingers was present. Pes

planus was found. The child had cryptorchidism. Central Nervous system examination revealed generalised hypotonia with preserved reflexes. NCCT showed cerebral atrophy. Cardiac auscultation revealed normal S1 and S2, with absence of any murmur.

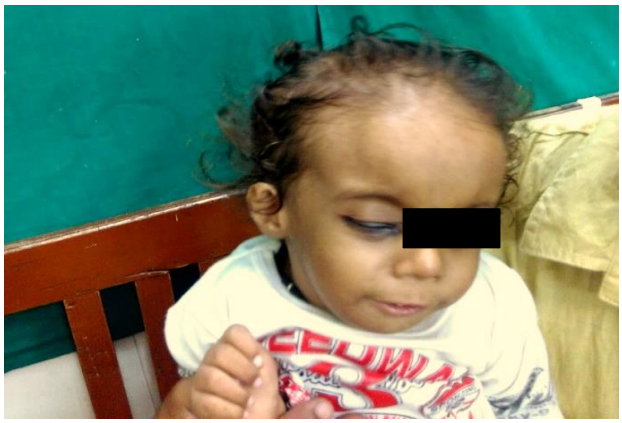

Figure 1: The child showing facial dismorphism (unilateral ptosis, low set ears, flat nose, retrognathia, prominent forehead).

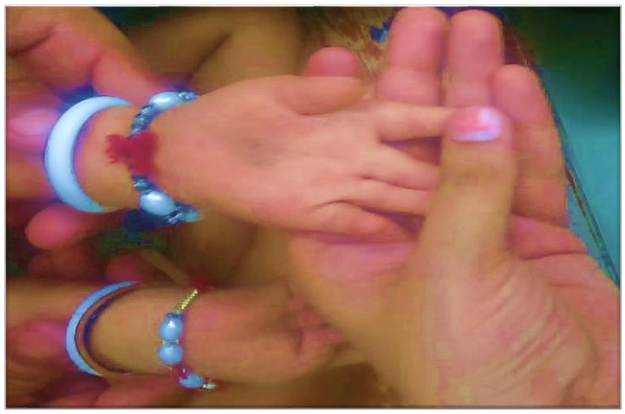

Figure 2: The photograph showing absence of palmar creases.

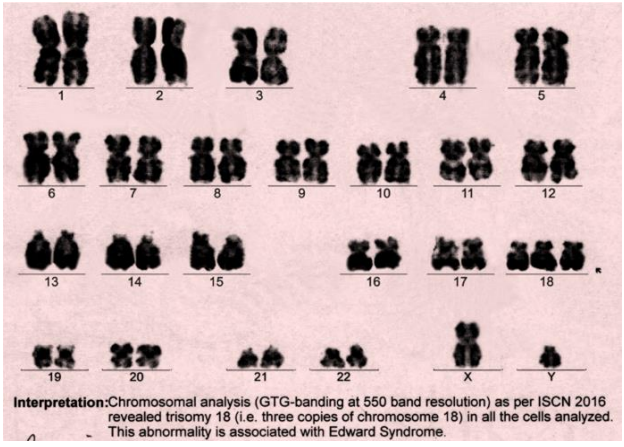

Figure 3: Karyotyping of the child showing three chromosomes at the level of 18th chromosome.

Rest of the system did not give any other abnormal finding. USG abdomen revealed non visualisation of testes in scrotum with no other abnormality of other organ. X-ray skull and vertebral column showed no defect in bony skeletal.

X-rays of limbs and joints did not show any limb defect or dislocation of hip. Echocardiography did not reveal any cardiac lesion. Karyotyping of the patient revealed three copies of chromosome 18 in place of two copies in all the cells (Figure 3).

DISCUSSION

Edwards syndrome was first defined in 1960 by Edward and his colleagues4 with the survival rate of 8.4% at the end of first year of life. The reported survival cases are of females5 more which is in contrast to present case. Advanced maternal age is thought to play an important role6, but in the present case the age of mother was found to be less than 25 years. This syndrome is characterised by small placenta, polyhydramnios and poor foetal movements with low birth weight weak cry and difficulty in swallowing in neonatal period. Typical muscular hypertonia and hypoplasia of the skin and subcutaneous tissues may manifest after one month 7 but the reported case showed generalised hypotonia.

Children with trisomy 18 may have microcephaly, short stature, mental retardation, cranio-facial abnormalities such as a small face, prominent occiput, micrognathia, cleft lip/cleft palate; upturned nose, narrow palpebral fissures, ocular hypertelorism, small mouth; limb abnormalities including overlapping fingers, camptodactyly, nail hypoplasia, narrow hips with limited abduction, short sternum, underdeveloped thumbs, absent radius, webbing of the second and third toes, clubfoot or Rocker bottom feet.[8,9] In 97% cases with trisomy 18, structural disorders are found at least in three organs; VSD has been reported in 67%, underdevelopment of reproductive organs in 50%, horseshoe kidney in 32%, omphalocele in 14%, diaphragmatic hernia in 11% babies and oesophageal atresia has been reported with a rate of 11%.[10]

In the reported case, facial dysmorphism, failure to thrive, mental deficiency, were present but in comparison to the usual presentation of syndrome with microcephaly, hypertonicity, deformities of fingers and limb were absent. Cardiovascular and renal abnormalities were also absent. Survival in trisomy 18 is related to the severity of congenital malformations and to some extent, the availability of paediatric care. Survival into childhood or beyond is rare. Thus, the case reported with this unusual presentation of trisomy 18 suggests the need for karyotyping and consideration of Edward syndrome amongst the rarity of genetic disorder.

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