

A Rare Case Report of Edwards's syndrome (Trisomy 18)

Amar Taksande^{1*}, Rewat Meshram², Amol Lohakare³ and Mrudul Mugawar⁴

¹Professor, Department of Pediatrics, Jawaharlal Nehru Medical College, Sawangi Meghe, Wardha, Maharashtra, India

²Associate Professor, Department of Pediatrics, Jawaharlal Nehru Medical College, Sawangi Meghe, Wardha, Maharashtra, India

³Senior Resident, Department of Pediatrics, Jawaharlal Nehru Medical College, Sawangi Meghe, Wardha, Maharashtra, India

⁴Resident, Department of Pediatrics, Jawaharlal Nehru Medical College, Sawangi Meghe, Wardha, Maharashtra, India

***Corresponding Author:** Amar Taksande, Professor, Department of Pediatrics, Jawaharlal Nehru Medical College, Sawangi Meghe, Wardha, Maharashtra, India.

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Abstract

Edward syndrome is a chromosomal condition in which all or a part of chromosome 18 is present in the cells of the body three times (trisomy) rather than the usual two times. Of all babies born with the extra copy of chromosome 18, 50% do not survive one week of age and 90% do not survive past the first year of life. The common clinical features were dysmorphic facies, microbrachycephaly, camptodactyly, clinodactyly, and abnormal dermatoglyphics, and mental handicap, cardiovascular or neurological abnormalities. Here, we report a case of Edwards's syndrome presenting with failure to gain weight, mental retardation and deformity of hand and foot.

Keywords: Edwards Syndrome; Camptodactyly; Mental Retardation

Introduction

Edwards's syndrome (Trisomy 18) is a genetic disorder caused by the presence of an extra 18th chromosome which results from non-disjunction during meiosis. It is the second most common autosomal trisomy, after Trisomy 21 (Downs Syndrome). It has been reported that the incidence of ES is 1 in 3000 to 7000 births and that survival beyond infancy is unusual. Less than 10% survive the first year [1-4]. It's three times more common in girls than boys. Most of fetuses with the syndrome die before birth and the causes include neurological and cardiovascular abnormalities. It is impossible to predict an exact prognosis during pregnancy or the neonatal period. Number of reviews present in the Western literature but only few cases were reported from Indian which stimulus us to report this case.

Case Report

A six years old female child presented in the pediatric out patient department with complaints of failure to gain weight and deformity of hand and foot. She was the only child born to nonconsanguineous marriage. Her father and mother were 31 and 28 years old respectively at her birth. She was born at preterm (8 months) by vaginal delivery at home, and birth weight was? 1.5 kg Detail record was not available. The antenatal period was reportedly uneventful. The motor and mental development was delay from birth. The child weighed 8.4kgs, height of 90cm and head circumference was 43cm. On general examination, triangular facies, prominent back portion of the head (occipital), low set ear, an abnormally small jaw, a small mouth with an unusually narrow roof, malocclusion of teeth, an upturned nose widely spaced eyes, squint, depressed nasal bridge, and eversion of upper and lower lips were present (Figure 1). Camptodactyly and absent palmar flexion creases of all the fingers (Figure 2) with hypoplastic nails were present. The metacarpophalangeal joints of the index, middle, ring and little fingers were flexed and had a reduced range of motion. Central Nervous system examination revealed hypertonia

with brisk reflexes in upper and lower limb. Cardiac auscultation revealed that S1 and S2 were normal and soft systolic murmur was present in the pulmonary area. No other abnormality detected on systemic examination. Abdominal sonography was normal. Renal function test, liver function test and thyroid function test were normal. Hearing and visual acuity were within normal limits. Echocardiography and Color Doppler does not detect any cardiac lesion. X-ray examination of the skull and vertebral column did not show any vertebral abnormality. Chromosome analysis of the peripheral blood cells revealed 47, XX, +18 chromosomes.



Figure 1: Childs with facial dysmorphism.



Figure 2: Absent planter flexion creases on the fingers of the child.

Discussion

Edward's syndrome (ES) was first recognized in 1960 by the discovery of the extra 18 chromosome in babies with a particular pattern of malformations. The general features constitute a low birth weight baby with a small placenta, polyhydramnios and poor foetal movements. In the neonatal period, the child presents with a weak cry and difficulty in swallowing. Typical muscular hypertonia and hypoplasia of the skin and subcutaneous tissues may manifest after one month [6]. 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; congenital heart disease (ventricular septal defect, atrial septal defect, patent ductus arteriosus), omphalocele, esophageal atresia, horseshoe kidney, hypertonia, short sternum and arthrogryposis [3-5]. Karunakaran and Pai [5] reported the first case in the Indian literature in 1967. In our case, the typical facial dysmorphic features, failure to thrive, feeding difficulty, mental deficiency, microcephaly, hypertonicity, flexion deformities of fingers were present. Cardiovascular abnormalities were 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 has been noted on rare occasions; when they have lived to the age of 15 or 19 years [7]. The diagnosis is confirmed by chromosome analysis. Many affected fetuses are detected by ultrasound scan during the second trimester of pregnancy and diagnosis can be confirmed antenatally by amniocentesis and chromosome analysis. Recurrence risk is low, except when the trisomy is due to a balanced chromosome rearrangement in one of the parents.

Conclusion

Edwards syndrome is a rare genetic disorder associated with multisystem involvement in babies that causes severe disability.

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