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A Case of Trisomy 18 Presenting with Severe Facial Abnormality and Phocomelia

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ABSTRACT An unusual phenotypic presentation was found in a trisomy 18 infant, who presented facial deformity, bilateral upper limbs phocomelia and talipes equinovarus. The report added phocomelia as a phenotype of trisomy 18, which was rare in previous literature.

INTRODUCTION

Trisomy 18 is the second (Crider et al. 2008) most common trisomy after trisomy 21. The most common dysmorphic features associated with trisomy 18 are well documented, including small premature appearance, a prominent occiput, clenched hands with a tendency for the index finger to overlap the third finger, the fifth finger to overlap the fourth finger, a short sternum, and low arch dermal ridge patterning on the fingertips. Radial aplasia usually associated with absence or hypoplasia of the first metacarpal and thumb, is well known in trisomy 18. Here we described a case of a male infant with trisomy 18 who was confirmed by peripheral blood karyotype with severe facial abnormality, bilateral upper limbs phocomelia and talipes equinovarus.

CASE REPORT

Our patient presented at birth with severe facial abnormality and phocomelia. There was no family history of any congenital anomalies on either side of the couple. Both parents were of Chinese descent. The mother was 37 years old and the father was 38 years old. The mother's antenatal history was significant only for mild anemia due to iron deficiency but otherwise was uneventful. She received only one ultrasound at 8 weeks of gestation, unscheduled antenatal care and missed the second trimester amniocentesis. This was the parents' second child following a first normal pregnancy and there was no history of smoking and alcohol and substance abuse during the pregnancy. The father was a cleaner who also had no significant past medical history.

The baby was born at 38^{+2} weeks of gestation via spontaneous vaginal delivery and was smaller than gestational age with regard to weight and length at birth. The baby (Fig. 1) present: male, birth weight was 2420g, length was 36cm, major findings included facial deformity, bilateral upper limbs phocomelia and talipes equinovarus. Multiple facial abnormality (Fig. 2) was noted to have shortening of palpebral fissure, ocular hypertelorism, flat nasal bridge, severe cheilopalatognathus, hypogenetic ears, short and wide neck. A congenital tetralogy of Fallot and maladjusted cardio-thoracic ratio were found by postnatal ultrasound. The infant was diagnosed trisomy 18 according to the analysis of the chromosome karyotypes 47,XY,+18 in peripheral blood and he died 3 days after birth. The parents agreed to donate the dead infant for medical research, which had been adopted by hospital ethics committee.

DISCUSSION

Trisomy 18 syndrome, also named as Edward's syndrome, was first described in 1960 by Edwards and colleagues (Edwards et al. 1960). The same year, David Smith and colleagues (Smith et al. 1960) reported the first patient in North America. The incidence of trisomy 18 is approximately 1 in 6,000-8,000 livebirths (Crider et al. 2008), and it is the second most common trisomy after trisomy 21. The

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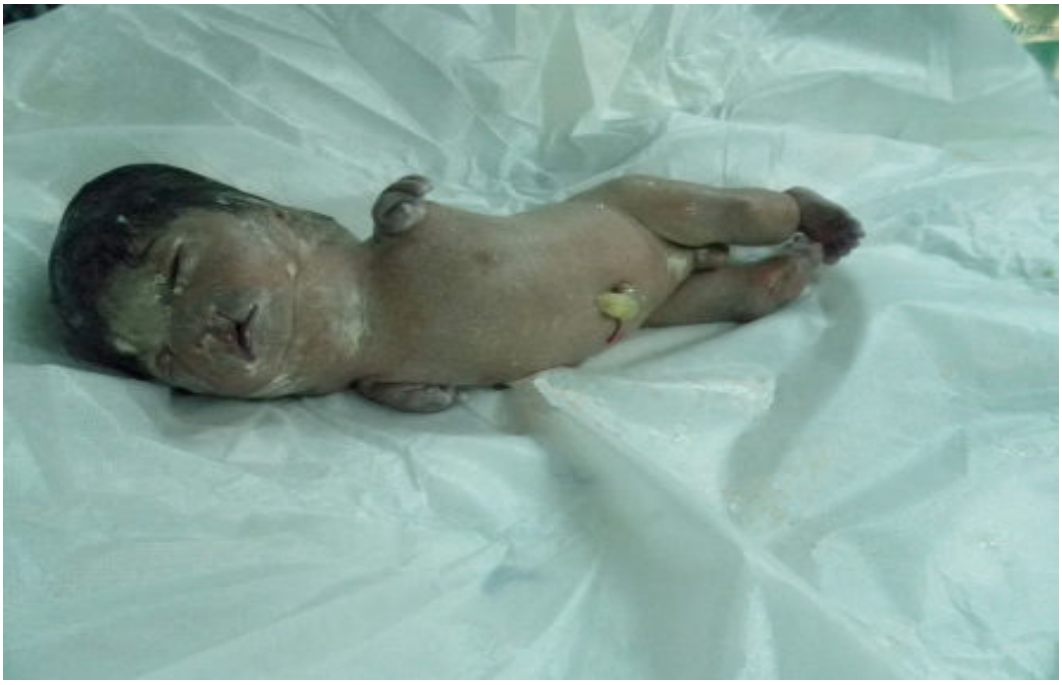

Fig.1. Whole body showing major findings including facial deformity, bilateral upper limbs phocomelia and talipes equinovarus.

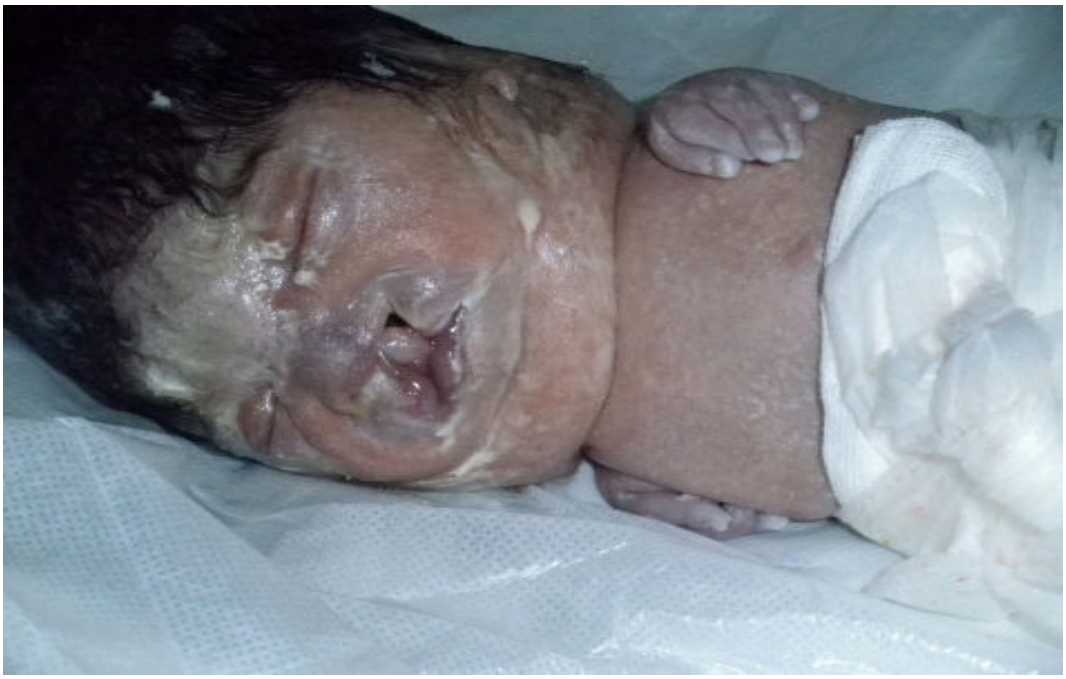

Fig. 2. Face with shortening of palpebral fissure, ocular hypertelorism, flat nasal bridge, severe cheilopalatognathus, hypogenetic ears, short and wide neck.

disease is associated with a high rate of intrauterine demise, with only 5% of the babies surviving until birth, with 95% of live-born infants with trisomy 18 die within 1 year (Crider et al. 2008).

The pathogen is nondisjunction of chromosome 18 during segmentation, which is frequently related to maternal senility and senescent oocyte. In this case the pregnant woman is 37 years old. Trisomy 18 is a multiple systematic malformation syndrome, the present case confirmed that. Typical abnormalities of trisomy 18 include abnormal appearances of the face, limbs and so on. Over half cases were complicated by limb abnormalities, those features in trisomy 18 include typical clenched hand with overlapping index finger, nail hypoplasia, radial aplasia, a short dorsiflexed hallux valgus and talipes varus. Four cases of trisomy 18 syndrome were reported by Christianson and Nelson (Christianson et al. 1984) with severe lower limb reduction anomalies: talipes equinovarus, and preaxial poly- or syndactyly. Trisomy 18 syndrome with cleft foot (Castle et al. 1988) was also reported. One case of trisomy 18 syndrome was strictly reported (Witters et al. 2008) with bilateral absence of radii, short humeri, absent right thumb and clefting of the hands in the upper limbs and bilateral absence of a distal bone (tibia or fibula) in the lower limbs. The upper arms deletion defects are more often present in the Roberts syndrome, the TAR syndrome and the phocomelus. The present case differs from those previously reported in bilateral upper limbs phocomelia, which is seldom found in trisomy 18. It is regretted that there is not X ray appearance of the arms.

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REFFERENCES

Castle D, Bernstein 1988. Trisomy 18 syndrome with cleft foot. *J Med Genet*, 25(8): 568-570.

Christianson AL, Nelson MM 1984. Four cases of trisomy 18 syndrome with limb reduction malformations. *J Med Genet,* 21(4): 293-297.

Crider KS, Olney RS, Cragan JD 2008. Trisomies 13 and 18: population prevalences, characteristics, and prenatal diagnosis, metropolitan Atlanta, 1994–2003. *Am J Med Genet Part A*, 146(7): 820-826.

Edwards JH, Harnden DG, Cameron AH, Crosse VM, et al. 1960. A new trisomic syndrome. *Lancet*, 1(7128): 787-790.

Smith DW, Patau K, Therman E, Inhorn SL 1960. A new autosomal trisomy syndrome: Multiple congenital anomalies caused by an extra chromosome. *J Pediatr,* 57: 338–345.

Witters I, Fryns JP 2008. Trisomy 18 presenting with severe limb deformations. *Prenat Diagn*, 28(6): 549-550.