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A Thai male neonate with absent radii: A rare feature of trisomy 18

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We describe here a male neonate with cytogenetically confirmed diagnosis of trisomy 18. Due to his clinical presentation with a rare feature seen in this syndrome, other disorders need to be included in the differential diagnosis. Besides absent radii with corneal opacities, this patient had other features including intrauterine growth retardation, prominent occiput, low-set ears, cleft palate, micrognathia, cardiac defect, hypospadias, imperforate anus, clenched hands and rocker bottom feet. These findings were consistent with trisomy 18.

This report emphasizes an importance of chromosomal analysis in patients with absent radii in addition to other anomalies.

Keywords: *Trisomy 18, Absent radii, Neonate.*

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กัญญา ศุภปีติพร, วรศักดิ์ โชติเลอศักดิ์. ผู้ป่วยไตรโซมี 18 ที่กระดูกเรเดียสไม่เจริญ. จุฬาลงกรณ์เวชสาร 2550 ม.ค; 51(1): 37 - 42

รายงานผู้ป่วยเด็กทารกชายที่มีท้ายทอยนูนชัด (prominent occiput), กระจกตาขุ่นขาวทั้งสองข้าง (corneal opacities), ใบหูเกาะต่ำ (low-set ears), เพดานปากโหว่ (cleft palate), คางเล็ก (micrognathia), หัวใจผิดปกติ, กระดูกปลายแขนท่อนนอกทั้งสองข้างไม่เจริญ (absent radii), อวัยวะเพศลักษณะ hypospadias, ไม่พบช่องทวารหนัก (imperforate anus), นิ้วมือทั้งสองข้างงอเกร็ง (clenched hands) และเท้าทั้งสองข้างเป็น rocker bottom ทั้งนี้ลักษณะที่จำเพาะที่สุดลักษณะหนึ่งคือ absent radii ซึ่งมีกลุ่มอาการที่ต้องให้การวินิจฉัยแยกโรคหลายโรค สำหรับผู้ป่วยรายนี้ได้ส่งวิเคราะห์โครโมโซม พบเป็น trisomy 18 ทั้งนี้ absent radii เป็นลักษณะที่พบได้น้อยมากใน trisomy 18 รายงานนี้แสดงให้เห็นถึงความสำคัญของการส่งตรวจโครโมโซมในผู้ป่วยที่พบมีการหายไปของกระดูกท่อนแขนส่วนนอก ร่วมกับความผิดรูปอื่น ๆ

คำสำคัญ : *ไตรโซมี 18, กระดูกเรเดียสไม่เจริญ*

Congenital anomalies involving multiple organs including the upper limbs can be found in several disorders with different etiologies. A chromosomal disorder is one of the most important causes.

We present a Thai male neonate with multiple anomalies consistent with several reported syndromes with one of the key features being absent radius. Chromosomal analysis confirmed a diagnosis of trisomy 18 in this patient.

Case report

The patient was the second child of a non-consanguineous 36-year-old mother and 38-year-old father. He had a healthy 10-year-old sister. His family history was unremarkable. He was born at 36 weeks gestation via cesarean section at King Chulalongkorn Memorial Hospital due to prolonged rupture of the membranes. His birth weight was 1,805 g (<3rd centile), length was 40 cm (<3rd centile) and head circumference was 37.5 cm (50th centile). His Apgar scores were 4 and 6 at 1 and 5 minutes, respectively. At birth he had weak cry and was hypotonic. However, no resuscitation was required. Clinical examination revealed prominent occiput, bilateral corneal opacities, low-set and malformed ears, cleft palate, micrognathia, systolic ejection murmur gr II/VI at LPSB, hypospadia, imperforate anus and rocker bottom feet. In addition, he had bilateral radial deviation with absent right thumb and clenched hands as shown in Figure 1.

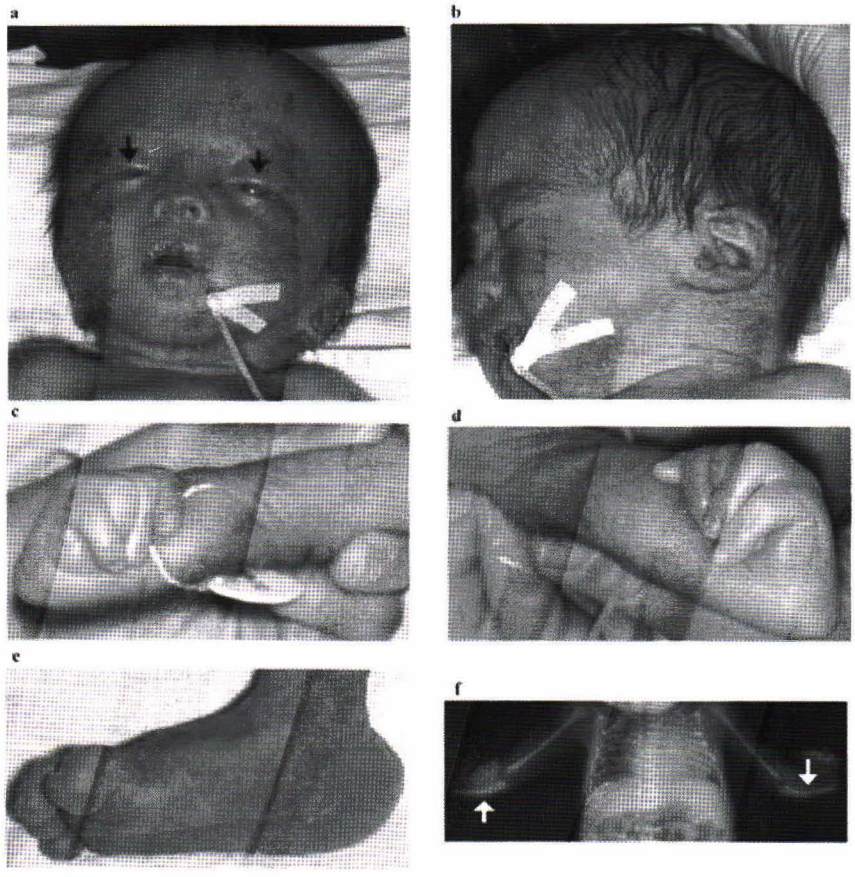

Figure 1. A patient with multiple anomalies: (a) bilateral corneal opacities; (b) low-set and malformed ear; (c) radial deviation of right hand and absent thumb; (d) clenched hand with radial deviation; (e) rocker-bottom foot; and (f) absent radii from X-ray.

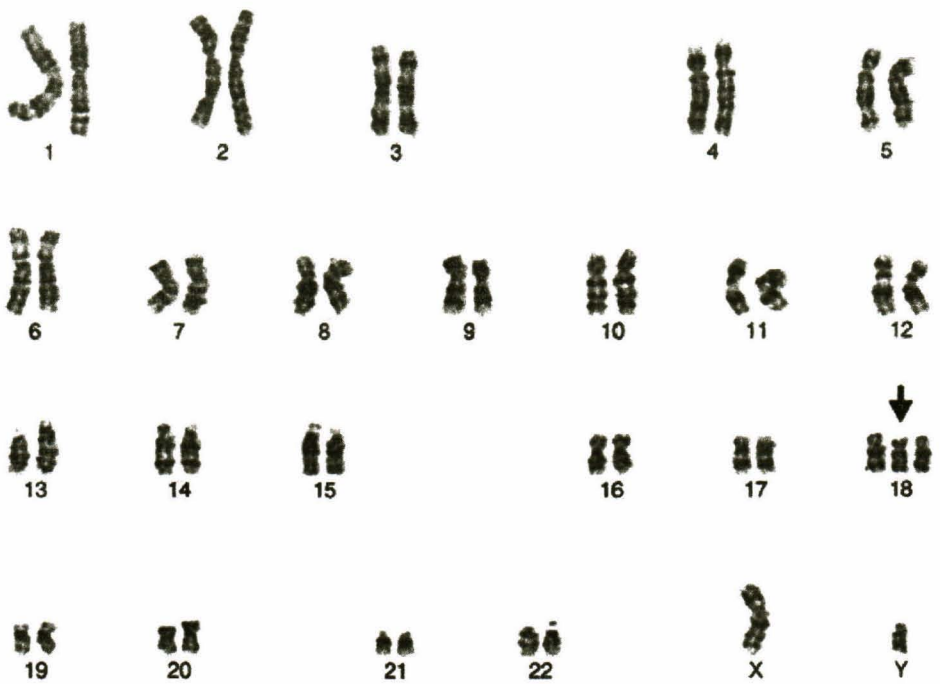

Figure 2. Karyotype of the patient showing trisomy 18 (46, XY, +18).

X-rays confirmed bilateral absent radii with absent right thumb. Brain ultrasound showed hydrocephalus. The renal ultrasound was normal. The patient died at 4 days old due to multiorgan failure.

Chromosomal analysis revealed 47, XY,+18 (Figure 2).

Discussion

Our patient had several clinical features consistent with trisomy 18 including intrauterine growth retardation, prominent occiput, low-set and malformed ears, cleft palate, micrognathia, cardiac defect, hypospadia, imperforate anus, clenched hands and rocker bottom feet. In addition, bilateral corneal opacities and absent radii which are rare features of trisomy 18 were found.

An absence of radius is rarely seen in trisomy 18. [1] Therefore, other congenital anomalies with upper limb reduction and radial aplasia need to be included in the differential diagnosis in order to make an appropriate management plan. Each disorder has its unique clinical features distinguishable from others. Table 1 shows syndromes with absent radius as a part of the disorders.

Trisomy 18 syndrome is the second most common autosomal trisomy after trisomy 21. [1] Its incidence ranges from 1 in 3,000 to 1 in 8,000 live births. [2,3] Previous studies have shown that 50 % of the affected infants die within the first week after birth and many of the remaining die within the following 12 months. Only 5 -10 % survive the first year as severely mentally defective individuals. [1] Recent studies have investigated the survival and natural history of trisomy 18 and revealed that almost all patients died within the first year due to severe malformations of the cardiovascular and/or central

Table 1. Syndromes with absent radius and other anomalies*

Syndromes
BALLER-GEROLD SYNDROME
THROMBOCYTOPENIA—ABSENT RADIUS SYNDROME
VENTRICULOMEGALY WITH DEFECTS OF THE RADIUS AND KIDNEY
LAURIN-SANDROW SYNDROME
DUANE-RADIAL RAY SYNDROME
SC PHOCOMELIA SYNDROME
RADIAL-RENAL SYNDROME
HOLT-ORAM SYNDROME
RADIAL APLASIA, X-LINKED

* http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM

nervous systems. The median survival was 4-6 days. The three most common structural defects were clenched hands, rocker bottom feet and low-set or malformed ears. [3,4]

An absence of the radius is among the abnormalities found in less than 10 % of trisomy 18 cases. [1,5,6] There was a recent report describing a fetus with trisomy 18 in which upper limb reduction was detected prenatally as an isolated defect during the second trimester screening by ultrasound. [7] It demonstrates the need for thorough ultrasound evaluation of the fetal hands as early as possible since upper limb defects can be the only abnormality of trisomy 18 found during that period. If there is clinical suspicion of trisomy 18, chromosomal analysis is required.

The majority of the cases have trisomy for all or a large part of the chromosome number 18. As for a full-blown case of trisomy 18, the result of faulty chromosomal distribution is related to advanced maternal age which is similar to other autosomal trisomies. Even though no adequate studies of recurrence risk have been performed, it is generally accepted that the recurrence risk would be lower than 1%.[1] Prenatal diagnosis by chromosomal analysis of fetal cells obtained from chorionic villus sampling or amniocentesis in high risk pregnant women can prevent the disease.

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