Clinical Brief

Distal Femoral Duplication and Fibular Agenesis Associated with Congenital Cardiac Defect

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ABSTRACT

A newborn, who had the congenital anomalies including protuberance on the right lower leg, bilateral equinovarus deformity of the feet, genu valgum with knee flexion deformity, syndactyly between the first and the second digit on the right, in addition with the absence of the fifth digit is reported here. Echocardiography revealed a secundum type atrial septal defect. The combination of these congenital defects associated with developmental anomalies of lower extremities. The clinical, radiological findings and pathogenesis of this lower extremity malformation is discussed. **[Indian J Pediatr 2010; 77 (2) : 210-211]** *E-mail: muratcak@hotmail.com*

Key words: *Embryogenesis; Lower extremity growth; Femoral duplication; Congenital heart defect*

Developmental anomalies of lower extremities are rare and heterogeneous malformations in childhood. According to hypothesis of Lewin and Opitz, two distinct developmental fields, the fibular and the tibial, control the development of the lower limbs.[1] We present the radiological and clinical findings of a case with unusual lower extremity malformations that supports the hypothesis of Lewin and Opitz.

REPORT OF CASE

A newborn male was delivered by cesarean section after 36 weeks of gestation and admitted to our neonatal intensive care unit for multiple congenital anomalies. Father and mother were 37 and 31 years old at the time of delivery, respectively. No parental consanguinity was present and family history was negative for birth defects. The mother denied having taken any drug or having had febrile illness during this pregnancy.

At delivery, his weight was 2800g (50-75%), length 46cm (25-50%) and head circumference 32cm (25-50%). Physical examination revealed the following abnormalities: protuberance on the right lower leg, bilateral equinovarus deformity of the feet, genu valgum with knee flexion deformity, syndactyly between the first and the second digit on the right, in addition with the absence of the fifth digit (Fig. 1). Other physical examination was normal.

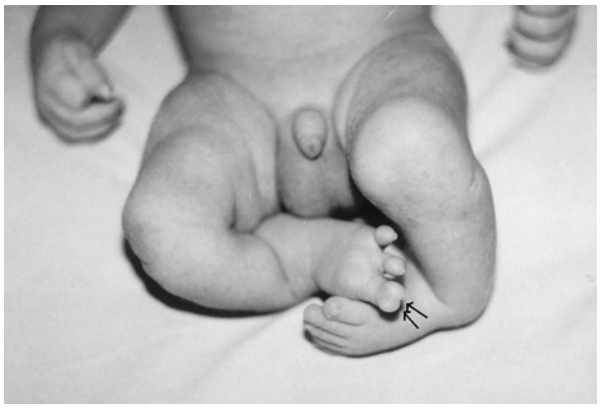

Fig. 1. Photographic image of the case showing lower limb anomalies. Note the hallucal syndactyly (black arrows) and absence of the fifth toe on the right foot.

Radiological examinations showed 11 pairs of ribs bilaterally and "butterfly" vertebrae at T7 and T10. Both fibular bones were absent. In the right lower limp; the femoral shaft was distally duplicated, and the distal femoral epiphysis and the tarsal bones were absent. Additionally, the metatarsals and phalanges of the fifth toe were absent. A protuberance originating from the distal femur was also noted on the left side. Both calcanei were ossified and appeared dislocated (Fig. 2).

Echocardiography revealed a secundum type atrial septal defect. Results of the cranial, lumbosacral

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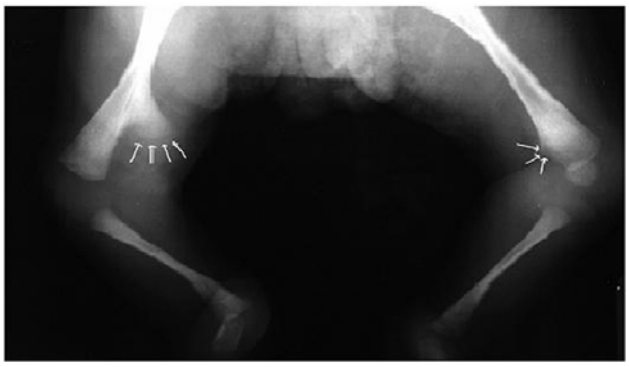

Fig. 2. Anteroposterior radiograph of the lower extremities showing distal femoral duplication (white arrows) and absence of fibular bone.

magnetic resonance (MR) were normal as was the abdominal and renal ultrasound. Lower extremity MR did not reveal any muscles and meniscus anomalies.

The karyotype was normal, and the diagnosis of small patella syndrome was excluded by the absence of TXB4 mutation (sequenced exons 1–8).

DISCUSSION

Congenital anomalies of the femur are rare malformations in childhood. Most of them are hypoplasia, bowing or proximal focal agenesis of the femur.[2] Congenital absence or total or partial duplication of the femur are considered rare defects.[3] In partial duplication, the duplicated bone can originate distally from the epiphysis or medially from the diaphysis. Congenital absence of fibula is the most common deficiency of the long bone deficiency.[4] It is generally associated with a variety of other lower limb abnormalities, including deformities of foot with the absence of one or more lateral rays, tibial bowing and femoral shortening.[1] Our case shows distally duplication of femur associated with bilateral fibular agenesis, hallucal syndactyly, agenesis of the right tarsal bone and fifth digit, and congenital cardiac defect.

Femoral duplication was firstly described by Erlich in 1885 in a case associated with ipsilateral tibial aplasia; and then it has been described infrequently and as an isolated entity in most cases.[3]

Our case supports the defects in both fibular and tibial developmental field defect bilaterally. Femoral duplication, syndactyly of the hallux, vertebral and rib anomalies are related with tibial developmental field defect. On the other hand, bilateral absence of the fibula, genu valgum, absence of the right fifth digit, right distal femoral epiphysis and tarsal bone, and joint dislocations represent fibular development defect. Cranial MR and abdominal ultrasound were normal in our case. We also performed lower extremity MR to demonstrate anomalies in the extraosseous structures of the lower limb. No muscles and meniscus anomalies were found.

Non-skeletal malformations were noted in four (0.8%) of the patients reviewed by Lewin and Opitz.[1] Two of them were neural tube defect; one was cardiac anomaly and the other renal anomaly. Cardiac defect was also noted in a case reported by Khoury *et al.*[5] Congenital cardiac defect may also be associated with other lower extremity malformation complex such as Gollop-Wolfgang complex, tibial agenesis-ectrodactyly syndrome, isolated tibial agenesis, and skeletal syndromes such as campomelic syndrome, Robinow syndrome and TAR syndrome.[6, 7]

Recently, Asomoah A *et al*[8] described a case with femoral bifurcation, fibular agenesis, congenital hearth and other multiple congenital anomalies associated with proximal chromosome 8q deletion and they suggest that a gene on chromosome 8q may be involved in lower limb development.

Many of the reported cases are sporadic in the literature. A few cases associated with autosomal recessive, autosomal dominant and X-linked transmission have been reported.[1] Normal karyotype and the absence of both consanguinity and positive family history in the present case support the sporadic feature of the disease. Nonetheless, a new dominant mutation can not be ruled out.

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