Case Report

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Radiology Section

# Bilateral Femoral Duplication and Fibular Agenesis Associated with Bilateral Lower Limb Ectrodactyly: A Rare Case Report

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## ABSTRACT

Limb deficiency disorders are rare, aetiologically heterogeneous skeletal dysplasias, they may occur as an isolated anomaly or as a part of syndrome. Femoral duplication is a rare anomaly which has been described as an isolated entity or in association with other congenital defects. We present the case of a 5-month-old male child with multiple congenital skeletal anomalies including protuberance on the bilateral lower legs, ectrodactyly of both feet, bilateral equinovarus deformity, genu valgum with knee flexion deformity and pre-axial polydactyly of the left hand. On detailed clinical evaluation of the patient, a provisional diagnosis of arthrogryposis multiplex congenital was made. Detailed radiographic imaging of the child was performed and multiple skeletal anomalies were identified.

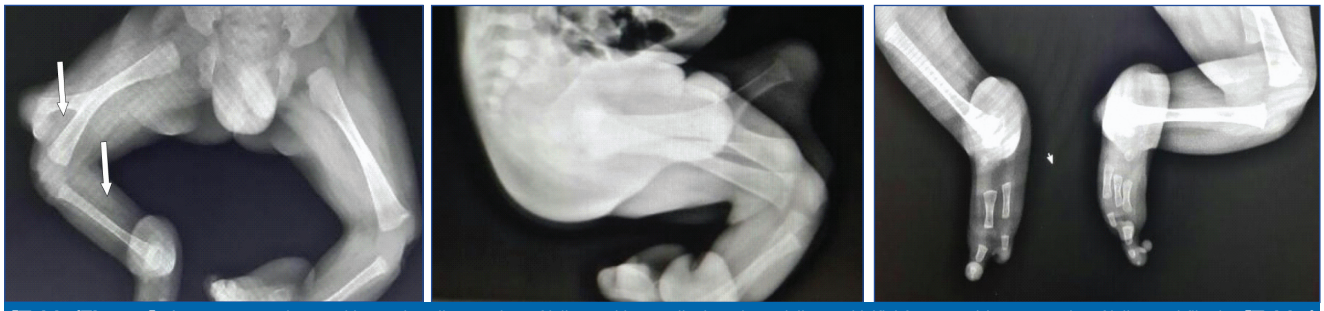
**Keywords:** Congenital disorder, Consanguineous marriage, Limb deformities

## CASE REPORT

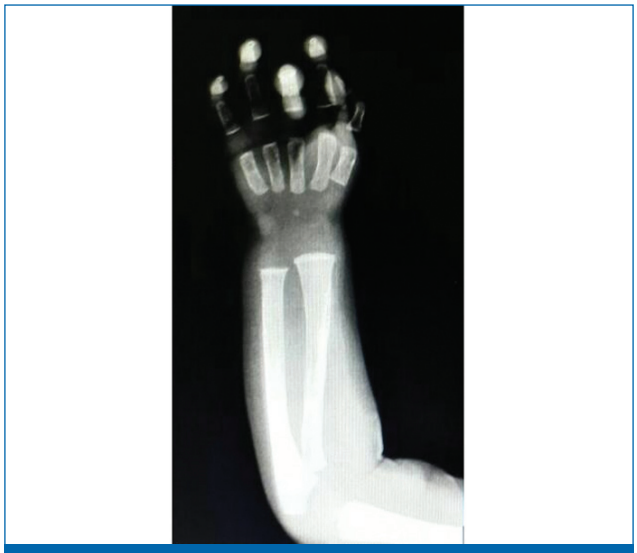
A 5-month-old male child was referred to our department for detailed radiographic imaging for multiple limb deformities. The child was born to a 26-year-old primigravida, full term by C-section and the post natal history was uneventful. The child was born of a consanguineous marriage and this was a sporadic case in the absence of any family history of similar malformations. There was no history regarding any exposure to radiation, prenatal teratogenic medications or infections during pregnancy. The mother did not smoke or consume alcohol during pregnancy. The child was breast fed with good appetite and cry, with no bowel and bladder problems, change in skin color or any cleft lip/palate. There was no history of any other comorbidities. Detailed clinical examination and radiographic evaluation of the child revealed a large protrusion that was present on the medial aspect of distal one-third of femur bilaterally (right>left) [Table/Fig-1]. Bilateral talipes equinovarus deformity was present [Table/Fig-2,3]. Genu valgum with flexion deformity of bilateral knees was also noted [Table/Fig-2].

On radiographic images, bifid femur was identified bilaterally with bilateral fibular agenesis [Table/Fig-4,5]. Ectrodactyly was

**[Table/Fig-1]:** Image showing a large protrusion present on the medial aspect of distal one-third of bilateral femur (right>left). **[Table/Fig-2]:** Image showing bilateral talipes equinovarus deformity with genu valgum with flexion deformity of bilateral knees. **[Table/Fig-3]:** Image showing bilateral talipes equinovarus deformity.



**[Table/Fig-4,5]:** Anteroposterior and lateral radiographs of bilateral lower limbs show bilateral bifid femur with agenesis of bilateral fibula. **[Table/Fig-6]:** Anteroposterior radiograph of bilateral feet shows ectrodactyly.



**[Table/Fig-7]:** Anteroposterior radiograph of left upper limb shows pre-axial polydactyly.

noted in both feet [Table/Fig-6]. In the upper limbs, only pre-axial polydactyly was noted in the left hand [Table/Fig-7].

A 2D echocardiography revealed no associated congenital heart defects. The sonography of abdomen and pelvis revealed no visceral or renal abnormalities. Other laboratory work-up of the patient was normal and the karyotyping was also normal (46XY). Treatment by surgical reconstruction was advised for the patient, but the parents did not give any consent for treatment.

## DISCUSSION

Skeletal dysplasias are generalised disorders of cartilage and bone which are associated with a abnormality in the skeleton [1]. Limb abnormalities can be seen isolated or may be detected as part of a known syndrome (associated with other malformations) [2].

Congenital anomalies of the femur are rare malformations that occur in childhood and most of them are hypoplasia, bowing or proximal focal agenesis [3]. Congenital absence of the femur or total/partial duplication of the femur is considered rare defects [4].

Ectrodactyly is the deficiency or absence of one or more central digits of the hand or foot which is also known as split hand/split foot malformation [5]. The main pathogenic mechanism is probably a developmental failure of the median apical ectodermal ridge in the growing limb bud [6]. It may be isolated or associated with other abnormalities.

Absence defects of the lower limbs are usually partial, and likely to be part of a more widespread multiple malformation complexes. Congenital fibular deficiency is more often sporadic and is the most common deficiency of the long bones [7]. Fibular deficiency is found in a variety of conditions, as part of a widespread skeletal disorder or a multisystem entity [8].

Asomoah A et al., described a case with femoral bifurcation, fibular agenesis, congenital heart and other multiple congenital anomalies associated with proximal chromosome 8q deletion [9]. Lower extremity malformation may also be associated with congenital cardiac defects such as Gollop-Wolfgang complex, tibial agenesis-ectrodactyly syndrome and isolated tibial agenesis [10,11]. However, in this patient, the karyotyping was normal and no cardiac defects were detected on echocardiography.

The aetiology of limb abnormalities is complex, it involves chromosomal abnormalities, single gene disorders, intrauterine factors, maternal and other various factors [12-15].

In this patient, there was no history of any chronic exposure to a toxic substance or infectious agent that could result in the disruption of normal skeletal development. Thus, these limb deficiencies could have been the result of spontaneous gene mutations.

## CONCLUSION

Bilateral femoral duplication associated with fibular agenesis is an extremely rare skeletal anomaly. Our case presented with multiple congenital skeletal abnormalities which couldn't be classified under any known syndrome. History of consanguinity may indicate a spontaneous gene mutation. However, normal karyotype and the absence of any positive family history may support the sporadic feature of the disease.

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