FEMUR – FIBULA – ULNA COMPLEX

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Introduction

We wish to report congenital limb abnormality – contralateral site deficiency which is rare anomaly and rarely reported.

Case Report

A 6 years old boy referred to our clinic for x-ray upper and lower limbs. Child was delivered preterm (35 weeks) with emergency cessarean section due to placenta previa and antepartum hemorrhage with congenital anomaly on antenatal ultrasound. Mother was anemic throughout pregnancy. On physical examination patient had absent left forearm, deficient left hand with presence of rudimentary thumb only, short right lower limb with deficient right lateral toes since birth. Mental status was normal, no facial or spinal abnormality noted.

Radiographic findings

Radiographs demonstrates: Agenesis of right fibula and the lateral two toes of right foot. (Fig. 1).
Abduction of the right foot and mild varus at the right knee joint. (Fig. 2).
The tibia is also short and shows anteromedial bowing. The right patella has not ossified. (Fig. 1).
The right femur is short and bowed. The shortening is more marked in the proximal portion of the femur and the femoral capital epiphysis has not formed. There is dislocation of the right femur with vertical and shallow dysplastic right acetabulum. There is formation of pseudoacetabulum in the right iliac bone. The right iliac bone also appears hypoplastic compared to the left. (Fig. 3). The left hemipelvis and left femur appears normal.

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Humeri were normal. Radius and ulna are absent on left side, complete agenesis of ulnar ray and gross deficiency of radial ray with presence of rudimentary thumb only. (Fig. 4a & 4b). The spine is normal (Fig. 4a).

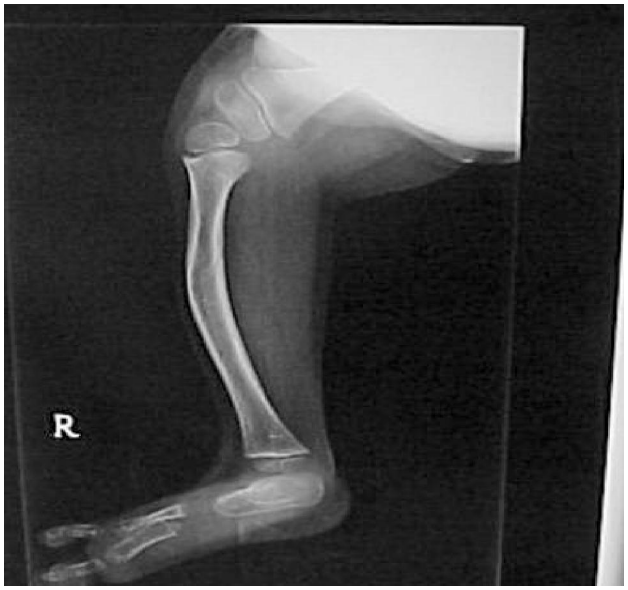

Figure 1

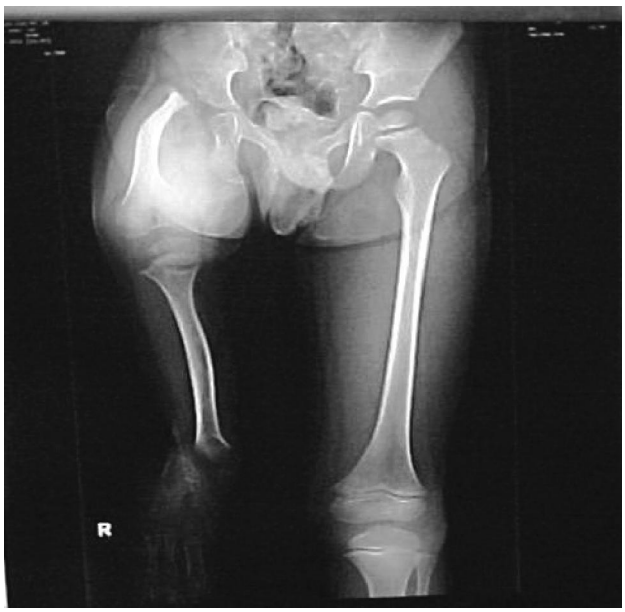

Figure 2

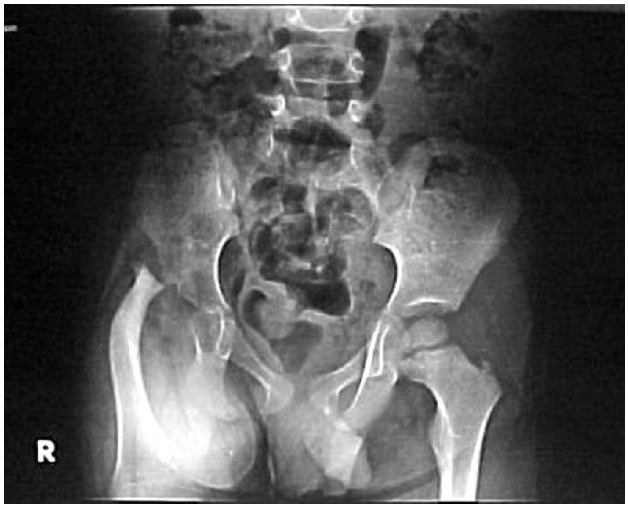

Figure 3

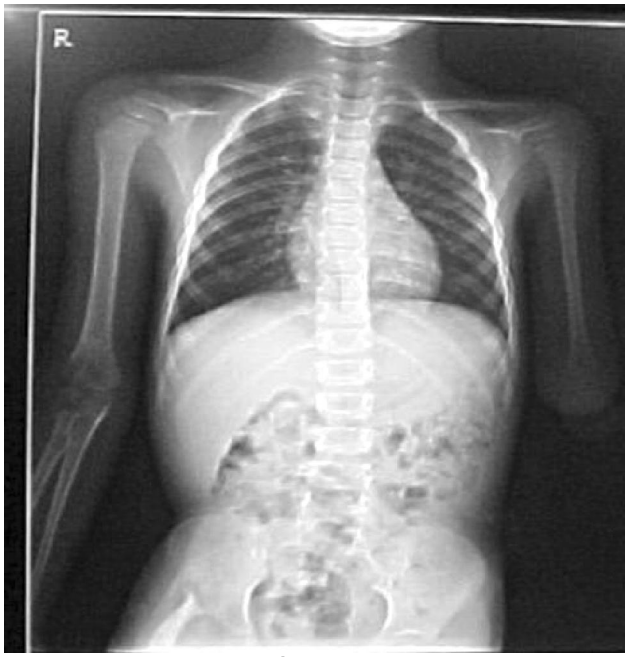

Figure 4a

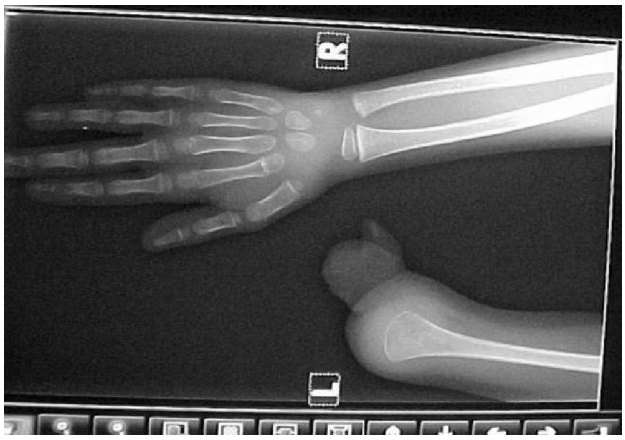

Figure 4b

Discussion

The femur fibula ulna (FFU) complex is a rare, mostly sporadic limb deficiency syndrome, consisting of a combination of femoral and fibular defects with malformation of the ulnar side of the upper limb. The typical defect of the femur can be designated as PFFD (proximal focal femoral deficiency) Upper limb defects, including Amelia (absence) of one arm, peromelia (peros:greek for mutilated) of the humerus, humeroradial synostosis and a defect of ulna or ulnar ray are usually a major constituent of this syndrome. Moreover, the upper limbs are more often affected than the lower limb. The lower limb deficiency may be located on the contralateral side of the upper limb defect. The right side upper limb is preferentially involved. Males are slightly more affected than females. The etiology is unknown and most cases of FFU complex are sporadic, with a negligible recurrence risk in siblings. Familial occurrence is very rare. There is no evidence for parental consanguinity or specific environmental causes or maternal / paternal age effect on FFU complex. However humeroradial synostosis with ulnar hypoplasia can be associated with fibular hypoplasia and this has been reported in ribs.

The differential diagnosis include other limb malformation syndromes, but the highly specific pattern of femoral defects associated with arm defects present in FFU complex is different from those seen in most other types of limb deficiency syndrome, so that there is virtually no overlap between these disorders.

FFU is not a lethal malformation. No primary treatment has been described.

Antenatal detection of major limb defects is possible by ultrasonography (as in our patient).

Conclusion

Due to absence of other visceral / spinal or neurological abnormalities, the patient has normal life uptill now (time of reporting) except the apparent physical abnormalities.

References

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