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FEMUR- FIBULA- ULNA COMPLEX

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Background: A 28-week-old female fetus was presented for a post-mortem radiographic examination. The pregnancy had been terminated after ultrasound examination (not shown) which demonstrated a small-for-date fetus with multiple limb malformations, including a short right arm with absence of the right fifth finger, absence of both thighs and absence of the left fifth toe. In view of these severe deformities, the mother had requested an abortion, and the pregnancy was terminated with the permission of the ethical committee of the hospital. Cytogenetic analysis indicated a normal female karyotype (46, XX). Anatomopathological examination confirmed the multiple limb malformations, but the internal organs were normal. Both parents were healthy non-consanguineous Caucasians.

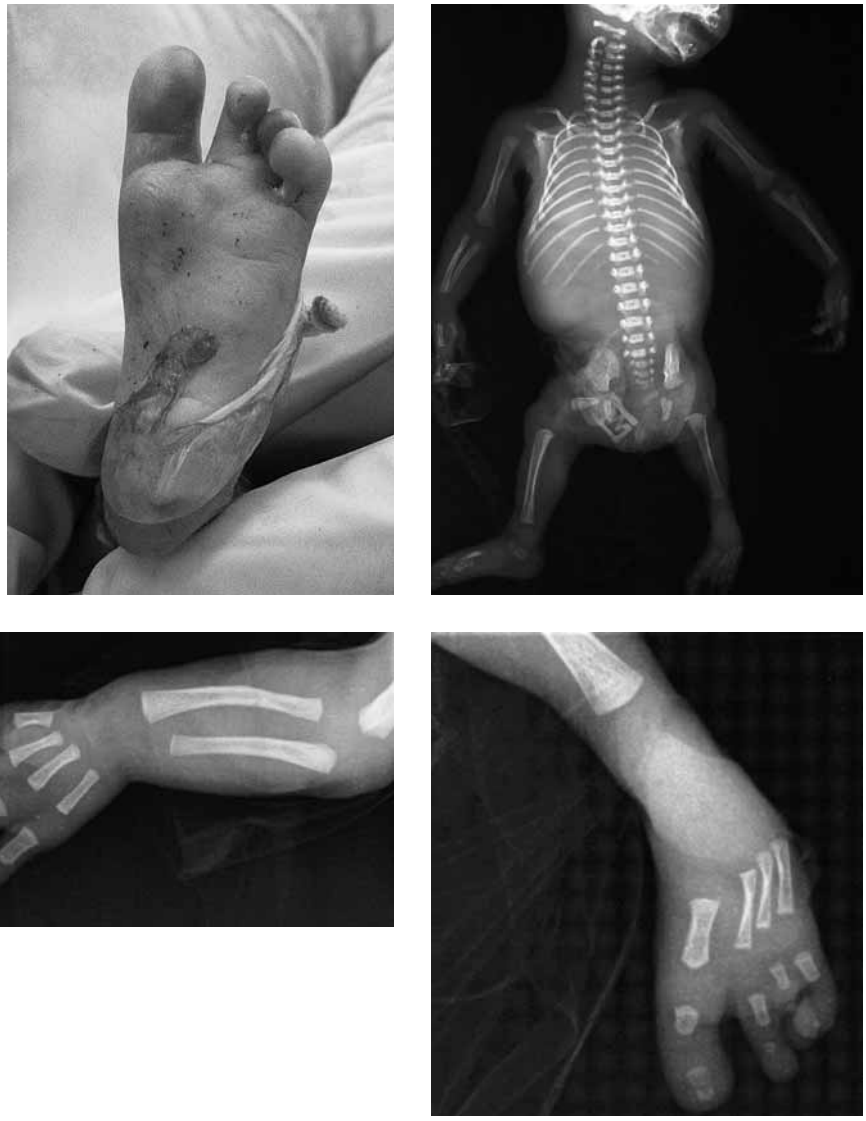

Fig. 1 | 2 / 3 | 4

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Work-up

Clinical photograph of the left foot (Fig. 1) shows absence of the fifth toe. On whole body radiograph (AP view) (Fig. 2), there is foreshortening of the right ulna and radius, with bowing of the right radius. The proximal part of the right femur is absent, with complete absence of the left femur and fibula. Spot radiograph of the right hand and forearm (Fig. 3) confirms the short ulna and radius, with bowing of the radius. Note the oligodactyly with absence of the fifth finger. Spot radiograph of the left foot (Fig. 4) demonstrates absence of the fifth ray. There is no ossification of the left calcaneus and talus.

Radiological diagnosis

Based on the association of defects of the femur and fibula with malformations of the upper limb, the diagnosis of *Femur- Fibula- Ulna (FFU) complex* was made.

Discussion

The Femur-Fibula-Ulna (FFU) complex is a rare, mostly sporadic limb deficiency syndrome, consisting of a combination of femur and fibula defects with malformations of the ulnar side of the upper limb (OMIM, Online Mendelian Inheritance of Man 228200).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 rays, are usually a major constituent of this syndrome. Moreover, the upper limbs are even more often affected than the lower limbs.

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 often affected than females.

The etiology is unknown and most cases of FFU are sporadic, with a negligible recurrence risk. 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 humero-radial synostosis with ulnar hypoplasia can be associated with fibular hypoplasia and this has been reported in sibs.

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

Furthermore, the absence of associated malformations of the internal organs on histopathological examination is a key feature in the differential diagnosis.

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

Antenatal detection of major limb defects is possible by ultrasonography. In this case, the findings were confirmed on conventional post-mortem radiography.

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