teogenesis imperfecta (Type II) are small for gestation, often stillborn or premature, have a relatively large head, deep blue black sclera, beading of ribs, virtual absence of skull bones and crumpled (concertina) long bones with or without camptomelia.

Genetic counselling to the parents, after due confirmation of the diagnosis is a must, as achondrogenesis is an autosomal recessive disorder, where the affected individuals are almost always born in only one generation of a family. Antenatal diagnosis with real time ultrasound is now possible to identify the various fetal skeletal dysplasias in early pregnancy, so that, those with the poor postnatal prognosis can be terminated(6).

Acknowledgements

The authors wish to acknowledge Mrs. Gladys D'Souza for all her help and the Department of Virology, NIMHANS, Bangalore for help in investigations.

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Proximal Focal Femoral Deficiency

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Proximal focal femoral deficiency (PFFD) which includes a congenital absence of proximal one third to two third of the femur often associated with other congenital anomalies, viz., absence of fibula, shortening of tibia, equinovarus foot, syndactyly, absence fourth and fifth rays, congenital dislocation of hip or oblique talus, has been studied by many authors, several of whom have proposed their own classifications. However, the most widely followed classification is by Aitken(1) based on the maturity of development of

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Received for publication: July 23, 1990; Accepted: April 16, 1992
acetabulum and femoral head. The absence of fibula unassociated with hypoplasia has been described by Caffey and in association with femoral hypoplasia by Acker(2).

We are reporting the radiological findings in three cases with classification and review of literature.

Case Report

Case 1: A 2-year-old male child was referred to the Radiology Department for the deformity of right leg and foot. There was no abnormality of any other system. None of the sibs or relatives had similar deformity. On roentgen examination (Fig. 1), there was hypoplasia of proximal and distal femur with a subtrochanteric varus deformity and sclerosis. The proximal end of femur was bulbous with a lucent rim between it and rest of femur. The lower end of femur was expanded and the femoral head was not seen in either of the acetabuli, which were well formed. There was complete absence of fibula with anterior bowing of tibia. The patient was classified as Aitken Class A and Robert’s type 2.

Case 2: A 1½-year-old male child presented with deformity of right lower limb since birth. Roentgen examination (Fig. 2) revealed deficiency of femur proximal to the level of trochanters. The proximal femoral end was bulbous in outline. There was no bony tuft extending proximal to it and there was absence of ossific nucleus of femoral head in the acetabular socket.

Fig. 1. Case 1 shows hypoplastic proximal and distal femur (Rt) with subtrochanteric varus deformity. Sclerosis of well formed acetabulum is seen with the absence of femoral head.

Fig. 2. Case 2 shows deficiency of femur with bulbous proximal end without a bony tuft. Ossific nucleus of femoral head is absent on Right side.

There was complete absence of fibula and tibia was bowed anteriorly. Ultrasonogram of hip (Fig. 3) as well as contrast arthrogram (Fig. 4) revealed well marked femo-
Fig. 3. Case 2. Ultrasonogram shows a well marked femoral head on right side.

Fig. 4. Case 2. Contrast arthrogram (right side) shows a femoral head outlined by contrast medium with synovial recess around it.

Fig. 5. Case 3 shows hypoplastic right femur with absence of femoral head. There is sclerosis and subtrochanteric varus deformity. There is absence of fibula with tibia bowed anteriorly.

The patient was classified as Aitken class C and Robert's type 3.

Case 3: A 1½-year-old male presented with deformity confined to right lower limb since birth. No other sibling was affected. On roentgenologic examination (Fig. 5), the right femur was hypoplastic with absence of femoral head. There was complete absence of fibula and the tibia was bowed anteriorly. There was no other abnormality in bones of foot. The patient was classified as Aitken class C and Robert's type 1.

The classifications in usage are briefly enumerated below:

1. Aitken's Classification:

This is the least complicated. Classes A and B are least severe(1). Class A: An adequate acetabulum and femoral head is present. A bony connection eventually
forms between femoral head and shaft. The point of connection is often pseudo-
arthritis which may subsequently ossify. **Class B:** An adequate acetabulum and femoral head. There is no bony connection between the femoral head and shaft. A small bony tuft is present at the proximal end of shaft. **Class C:** Marked dysplasia of acetabulum, femoral head does not form, distal femoral segment is very short and has an ossified tuft of bone at its proximal end. **Class D:** The acetabulum and femoral head are both absent. There is bony tuft at the proximal end of deficient femoral shaft.

2. **Fixen and Lloyd Robert’s classification(3):**

   **Type 1:** Bulbous proximal end of femoral shaft. A zone of sclerosis at midshaft level and angulation at the area of sclerosis. **Type 2:** Zone of sclerosis at the proximal end of deficient femoral shaft which is not bulbous but rather has an ossified tuft separated from the rest of shaft by a lucent line. **Type 3:** Similar to type 2 but lacks the ossified tuft.

3. **Functional classification by Ali Kalamchi(4):**

   **Type 1:** Short femur with good hip joint, the so called miniature femur. **Type 2:** Congenitally short femur with coxa vara. **Type 3:** Short femur with proximal deficiency, a well defined hip joint. **Type 3A:** The defect may ossify; **Type 3B:** The defect never ossifies. **Type 4:** Dysplastic distal femoral segment with no hip joint. **Type 5:** Total absence of femur.

**Discussion**

Golding(5,6) in 1939 and 1948 proposed that short femur of micromelia and deformity called congenital coxa vara are variation of same. Amsutz and Wilson(7) in 1962 discussed the cases of dysgenesis of proximal femur in which the essential components were femoral shortening and coxa vara.

Amsutz(9) has shown a constant growth ratio between normal and abnormal femur by radiography. Schatz(8) believed that tuft of bone mentioned in both the classifications (Aitkens and Fixsen Lloyd Robert’s) is poorly ossified femoral head and this can be verified by arthrography which has a role in any case of PFFD, if femoral head does not appear by one year of age.

The problems encountered in applying the classification of PFFD are due to normal failure of femoral head to appear till one year of age and difficulty in assessing the development of acetabulum which is a clue to the presence of normal femoral head even when it is not seen on radiograph. Further, a stable hip on early film might later become unstable(8) due to dissolution of pseudoarthritis or progressive coxa vara(8). Fixsen and Lloyd(3) have pointed out that upward and outward migration of femoral head is an indicator of unstable condition.

Fixsen and Lloyd(3) suggested a relationship between severity of femoral shortening and hip instability. Shelf index, acetabular index and acetabular dysplasia have been used as predictors of pelvic stability at maturity. Hillman et al.(10) found that correct classification was possible in 86% of cases by using these parameters.

The absence of fibula without deficiency of femur was described by Caffey(11). Acker(2) and Vyas and Gupta(14) described absence of femur associated with absence of fibula in a two-year-old child. In the present series absence of fibula is reported as a component of PFFD.
Aitken(1) did not include congenital coxa vara as a part of PFFD while Amsutz(8) included it in his classification. Important points for prognosticating hip stability are extent of limb discrepancy and location of apex of varus deformity. The type of deficiency at skeletal maturity may differ from that initially assessed.

Infantile coxa vara is separate from this entity as this is due to rotatory malalignment rather than defect of bone, does not present at birth and has a vertical lucent line running through femoral neck, besides it has a normal length of diaphysis unlike PFFD. Neglected congenital dislocation of hip might also be confused with it but has normal subtrochanteric region unlike PFFD.

Hillman et al.(10) used MR imaging in one case to demonstrate the unossified portion of femoral head obviating the need of other invasive studies.

There is every reason for attempting to make early prenatal diagnosis of the defect and towards this ultrasonography can be helpful(15). As such usefulness of a short femur in utero detection of skeletal dysplasias is well recognized and has been reported besides others by Kurtz and Needleman(16).

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