Congenital bifurcation of the femur with aplasia of the tibia

A case report

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Summary

Congenital bifurcation of the femur in association with aplasia of the tibia has been reported in only about 20 cases. One case is reported, the physical findings in this case and in others are described, and the important prognostic features are outlined. Amputation at knee joint level is a common outcome.

Bifurcation of the distal femur with ipsilateral aplasia of the tibia was first described by Ehrlich in 1885. Liepman reported a similar case in 1906. To date, about 20 cases of femoral bifurcation and about 200 cases of tibial aplasia have been reported. From a review of the literature it is apparent that the two conditions are often concomitant, while other associated abnormalities are common. These include hypoplasia of the ipsilateral femur, diplodia, polydactyly, duplication of the fibula and abnormalities of the upper extremities including the hand.

Bifurcation of the femur may occur distally, as in the case reported here, but duplication has been reported at various levels, sometimes including the entire limb.

Experimental evidence has shown that the initiation of the abnormality is usually in the mesodermal anlage of the femur. In our case there was failure of the proximodistal differentiation sequence. Some workers have postulated that the bifurcated portions of the femur represent an ectopic anlage of the tibia, and they support their claim with the presence of a fully developed pes anserinus implanted on the femoral segment.

Franz and O'Rahilly offered a classification of congenital skeletal limb deficiencies in 1961, followed by the classification of Jones et al. in 1978, for congenital aplasia of the tibia. In terms of the latter, our case may be classified as a Jones et al. type IA, with a poor prognosis indicated by the skin dimples over the knees and small or absent epiphyses of the distal femur (Fig. 1). The absence of the patella and the patella tendon is considered by Jones et al. to be the ultimate criterion of a poor prognosis. In this group amputation at the knee is mandatory.

Reconstructive procedures are sometimes possible, and Brown has described centralization of the fibula in the intercondylar fossa of the femur. This is possible only when a functional knee joint may be the result.

Fig. 1. Classification of congenital aplasia of the tibia (Jones et al.).

If the foot is present, amputation should be deferred and the appendage used to secure the plaster of Paris cast which is applied below the knee. Later, a Syme's amputation will enable the fitting of a satisfactory below-knee prosthesis. Excellent results have been reported with this procedure.

Case report

A healthy Black girl, aged 3 months, presented at Kalafong Hospital, Pretoria, in April 1982. She was the third child of a healthy, 36-year-old mother. The pregnancy and delivery were normal, the latter somewhat precipitate. A 'cross-legged' deformity was noted at birth. The child was healthy, but had an umbilical hernia and grossly deformed legs (Fig. 2).

There was wide abduction with flexion and external rotation at both hips. There was bifurcation of the lower end of the right femur and absence of tibia on both legs. There were large dim-
Fig. 2. Appearance at 3 months of age. There is wide abduction of the hips, flexion deformity of both knees, and gross deformity at below-knee level.

Fig. 3. Radiographs showing bifurcation of the distal femur on right, absence of both tibiae, and a well-developed fibula on both sides. There is a rudimentary epiphysis of the lower and right femur.

Discussion

This anomaly, although rare, has been well described and classified. The absence of a quadriceps mechanism is regarded as an indication for amputation. In less severe cases the limb might be preserved.

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