Proximal Focal Femoral Deficiency
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History
Newborn with limb anomalies.

Diagnosis
Proximal focal femoral deficiency with contralateral fibular hemimelia.

Discussion
Proximal focal femoral deficiency (PFFD) represents variable dysgenesis of the proximal femur. Most cases are isolated but PFFD can occur in conjunction with caudal regression or in infants of diabetic mothers. Most are unilateral. PFFD results in average shortening of about 60%. Also commonly seen is ipsilateral tibial shortening, equinovarus deformity and anomalies of the lateral digits. The underlying pathogenesis is related to altered proliferation of chondrocytes in the proximal femoral physis.

Prognostic indicators include 1) presence or absence of the femoral head, 2) normal or dysplastic acetabulum, 3) absence or length of distal femoral ossification, and 4) presence or absence of bony connection of the femoral head and more distal femur. Ultrasound or MR is helpful to better define the femoral anatomy.

PFFD is frequently seen in association with ipsilateral fibula hemimelia (50% of cases). This case is unusual in that the ipsilateral fibular hemimelia is minimal (if at all) but the contralateral fibular hemimelia is complete.

Findings
CR-Left femur is nonossified and short. The proximal left fibula is subtly thin. The right tibia is bowed and the fibula is nonossified.
US-The left femur is completely cartilaginous but has normal hip and knee articulations. The right fibula absent.

Reference
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