Case reports

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Monozygotic twins concordant for congenital short femur

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SUMMARY We report concordant male monozygotic twins with congenital short femur (proximal focal femoral deficiency) and discuss the aetiological implications. Coincidentally, they and their father have benign familial macrocephaly.

Congenital short femur (proximal focal femoral deficiency, PFFD) is an uncommon malformation which ranges in severity from mild hypoplasia with shortening and bowing to total absence of the femur. The lesion may be bilateral and in most cases there are other limb malformations, especially absence of the ipsilateral fibula and lateral foot rays (femur-fibula or femur-fibula-ulna complex). Rogala et al. found an incidence of congenital short femur of 1 in 52 029 in Edinburgh and there have now been more than 350 cases reported, mostly in orthopaedic publications. Familial recurrence, however, appears to be exceptional and thus we wish to report concordant monozygotic twins.

Case report

These male twins were the result of the first pregnancy for a non-consanguineous, normal Scots couple who had no family history of skeletal abnormalities. The father was 45 years and the mother was 38 years old at the time of delivery. The pregnancy had been uneventful and no medications or alcohol had been taken. Delivery was by elective Caesarian section at 39 weeks in view of suspected cephalopelvic disproportion. Birth weights were 3 kg (twin 1) and 2.8 kg (twin 2). Histological examination revealed a diamniotic monochorionic placenta, thus confirming monozygosity.

At 1 year of age shortening of the right leg in twin 1 was noted and a radiograph showed shortening of the right femur with angulation at the midpoint and coxa vara (fig 1). Twin 2 was considered normal at this stage but no radiograph was performed. Examination of twin 1 at 17 months of age revealed a standing height of 77 cm (10th centile) with 5.5 cm of true shortening of the right leg. Abduction of the right hip was limited but other movements were normal. Examination of twin 2 at this time revealed a standing height of 80 cm (25th centile) with 2 cm of true shortening of the right leg. Joint mobility was normal but a radiograph showed shortening of the right femur and coxa vara (fig 2). Both twins were otherwise normal except for macrocephaly (twin 1 OFC 52.5 cm at 17 months, twin 2 OFC 55 cm at 17 months), which was believed to be due to benign familial macrocephaly in view of the father's head

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of the left pectoralis major muscle, hypoplasia of the left arm, symbrachydactyly, and ipsilateral coloboma of the optic disc. This is the first report of the association of these two congenital anomalies.

**References**


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