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*¹ 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 exceptional²⁻⁴ 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

FIG 1 Twin 1 aged 1 year.

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FIG 2 Twin 2 aged 17 months.

circumference of 58.5 cm and the absence of neurological findings.⁵

Discussion

With the present report there are now four instances of familial recurrence of congenital short femur. Ring² described a pair of concordant twins with congenital short femur in 1959, but no comment was made about their zygoty. Kelly³ noted that a third degree relative had a curved femur, and in his

comprehensive family study Hamanishi⁴ found only one familial recurrence in first cousins among 56 families studied. In this study,⁴ no raised parental ages were found and only one set of parents was consanguineous. Furthermore, three sets of discordant twins of uncertain zygoty have been reported.^{4,6}

With the exception of thalidomide, which is usually associated with more severe femoral defects, no consistent environmental factors have been identified. Unifactorial inheritance is most unlikely and the observed patterns of familial recurrence and twin concordance favour multifactorial inheritance for simple femoral hypoplasia.

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Poland syndrome associated with 'morning glory' syndrome (coloboma of the optic disc)

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SUMMARY A 12 year old girl with the Poland syndrome and the 'morning glory' syndrome is described. The patient presented with absence

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.

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