BILATERAL CONGENITAL APLASIA OF THE FEMUR

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In 1853 Ellis published "An account of an instance of remarkable deformity of the lower limbs." Since then accounts of congenital deformities of the femur have appeared sporadically (Maguire 1914, Theobalds 1914, Fairbank 1926, Veitch 1926, Langston 1939, McFarland 1950, Acker 1959, Frantz and O’Rahilly 1961). Most have concerned cases of unilateral hypoplasia of the femur, with or without congenital coxa vara. Cases of bilateral affection have been few (Ellis 1853, Langston 1939, Katdare 1945, Ring 1961, Amstutz and Wilson 1962), and in most of them it is questionable whether the defect should be classified as a simple hypoplasia. A study of the papers and the radiographs published with them reveals an associated element of congenital coxa vara. In most of these accounts the condition as seen in children is described, and there is no description of the condition of the patients in adult life. In this account an unusual case of congenital and symmetrical aplasia of the femora in an adult is described.

CASE REPORT

A man aged fifty attended with a complaint of pain in the upper part of the right lower limb gradually increasing over two years, and occasional pain in the back.

The patient said that from birth he had had short lower limbs. He could not squat or sit cross legged and had to defaecate standing, but otherwise the deformities caused no severe disability. Before the onset of the pain he could walk four or five miles daily with a heavy bundle on his head and for the past ten years he had earned his living as a broom vendor.

His three brothers and one sister and his parents were normal. The patient lived a normal family life though he had no children.

Examination—The patient was stout but short—only three feet ten inches in height. The trunk was out of proportion to the lower limbs. He had a waddling gait with slight forward propulsion and walked fairly fast. The thighs were almost absent, so that the legs seemed to articulate directly with the pelvis. There were rudimentary buttocks (Figs. 1 and 2). In the upper lateral part of each buttock a bony prominence about one inch in diameter could be felt. This seemed to be the upper end of the femur. These bones moved on the pelvis and formed rudimentary thighs which joined the legs at an angle of about 100 degrees.

The power of the muscles of the legs was normal. The trunk and upper limbs were very well developed. There was a slight thoraco-lumbar scoliosis to the left, with exaggerated lumbar lordosis and gross restriction of all spinal movements. The length of the body from the vertex to the umbilicus was the same as that from the umbilicus to the middle of a transverse line joining the heels. The lengths of the right and left lower limbs from the anterior superior iliac spine to the medial malleolus were respectively nineteen and nineteen and a half inches. The distance from the anterior superior iliac spine to the bony prominence of the upper end of the tibia was three and a half inches on each side.

There was marked tenderness over the postero-lateral aspect of the upper end of the right tibia.

The patient could only sit on the ground with his lower limbs flexed at the hip, the right laterally and the left medially rotated and both deviated 45 degrees to the left.

Radiological examination—Radiographs showed extreme hypoplasia of the femora, symmetrical on the two sides. Each femur had a hypoplastic head, neck and trochanters. The shaft of each
The patient, seen from the front and from the side. Note the disproportion between the trunk and the lower limbs, the absence of the thighs and the rudimentary buttocks.

Radiograph showing the rudimentary femora and the plane joints between the femora and the tibiae.
femur was only about two inches long. The lower end was of normal width but showed no condyles or intercondylar notch. The upper surface of each tibia was flat and was without condyles so that it formed a plane joint with the femur. The acetabula were very shallow and there was hypoplasia of both iliac bones. Both hips were dislocated, the femoral heads being high up on the ilia. The patella was absent on both sides (Fig. 3).

Arteriographs showed a normal disposition of the vessels in the area.

Course—After the arteriography the patient was rapidly relieved of his pain and was able to resume his normal work. We believe that this relief was produced by the organic iodide used in the arteriography. However, it is expected that the degenerative changes in the right knee joint—mainly affecting the upper end of the tibia—are likely to lead to a recurrence of pain. In that case an arthrodesis of the knee with some form of internal fixation will have to be considered (Seddon 1962).

**COMMENT**

Hypoplasia of the femur is uncommon. Generally one side only is affected. Bilateral affection is rare and is usually asymmetrical. In Ellis's (1853) case the left femoral shaft was completely absent, while on the right the femoral and tibial elements had fused to form a composite bone (Figs. 4 and 5). The anterior muscles of the thigh were underdeveloped and there were associated arterial anomalies. In Katdare's (1945) case both femoral shafts were symmetrically absent. The femoral condyles—concave proximally—articulated with the underdeveloped femoral heads, which had fused with the ischia (Fig. 6). The patient had a left claw hand. In other cases associated abnormalities such as absence of the fibula and patella, syndactyly and arachnodactyly have been reported (Manohar 1939, McFarland 1950, Acker 1959, Frantz and O'Rahilly 1961, Ring 1961).

There is in fact a very gentle gradation from simple hypoplasia to complete aplasia of the femur (Golding 1939, 1948, Burrows 1950, Frantz and O'Rahilly 1961, Gordon 1961, Amstutz and Wilson 1962), but it is convenient to regard the whole condition as a distinct clinical entity that appears in several forms (Seddon 1962).
Simple femoral hypoplasia affecting the shaft and upper end—In this type the lower end of the femur is relatively well formed but the shaft and upper end are represented by a short bar of bone tapering upwards to a small and irregularly shaped head. The case reported here falls into this category (McFarland 1961, Gordon 1962).

Aplasia of the femoral shaft—The head and the lower end of the femur are relatively well developed but the shaft appears to be entirely absent (Van Nes 1950). Both limbs in Katdare's (1945) case and the left limb in Ellis's (1853) case showed this type of abnormality.

Femoro-tibial fusion—The femur and tibia are fused to form a composite bone, with small outlying knobs representing the head and trochanters of the femur. In Ellis's (1853) case the right lower limb showed this type of deformity.

Femoral hypoplasia with coxa vara—This is the abnormality in most reported cases of femoral hypoplasia. There is a short bony femoral shaft, the upper end of the femur being represented by a cartilaginous mass arising from the medial side of the femoral shaft and projecting inwards at an angle of 90 degrees to the femur. A condition of congenital coxa vara is thus produced. The presence of the cartilaginous precursor of the upper end of the femur can often be demonstrated clinically by the presence of a mass in the femoral triangle, and arthrography outlines the head and neck. Exploration will confirm the presence of the cartilaginous upper end of the femur (Amstutz and Wilson 1962).
condition is recognised in childhood and if observation is continued until maturity the true nature of the abnormality becomes evident (McFarland 1950, Speed and Knight 1956). The appearance of this type of abnormality in maturity is shown in Figures 7 and 8 and should be contrasted with the appearances in our case of bilateral affection.

**SUMMARY**

1. An unusual case of bilateral and symmetrical congenital aplasia of the femur in a man of fifty is described.
2. The literature on the condition is reviewed and discussed.

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**REFERENCES**


