CONGENITAL ABNORMALITIES OF THE FEMUR

BY

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Congenital defects of the femur vary from simple hypoplasia of the bone to complete absence. Classification of these defects has been suggested by Nilsonne (1928) and by Mouchet and Ibos (1928), but neither has met with general acceptance. In more recent years Golding (1939, 1948) has demonstrated the close association of the short femur with congenital coxa vara, and has emphasized that these are variations of the same underlying abnormality. The clinical distinction between the various types of femoral defect is important as a guide to the prognosis of limb development.

From an examination of patients with congenital abnormalities of the femur the following classification is suggested:

1. Simple femoral hypoplasia.
2. Congenital coxa vara.
3. Femoral hypoplasia with coxa vara.
4. Pseudarthrosis of the femur.
5. Hypoplasia of the lower femoral epiphysis.
6. Complete absence of the femur.

Although these patients commonly have other skeletal defects, the major congenital abnormalities affecting the nervous system, or in the chest and abdomen, appear to be rare. Their span of life is thus rarely affected, and detailed autopsy or pathological examination of the limb is rarely possible. From the clinical examination of these patients, however, and from serial radiographs, much information can be obtained about the clinical course of the limb abnormality, and the nature of the underlying disturbance of growth.

Clinical Material

The records of the Royal National Orthopaedic Hospital and The Hospital for Sick Children for the past 10 years have been examined, and patients with this diagnosis have recently been reviewed. With the addition of one case from Guy’s Hospital, 38 examples of this defect have been collected, 19 of these have been reported in detail elsewhere (Ring, 1959) under the title of simple femoral hypoplasia, and will only be considered briefly. A true indication of its incidence is difficult to obtain, but it appears to be the commonest congenital defect causing major abnormalities of limb growth.

Clinical Features

There is no evidence that this is a familial disorder, and careful inquiry of the parents has revealed no evidence of other congenital disorders within the immediate family. The history of the pregnancy and delivery has failed to indicate any significant infection or abnormality at this time. In most patients the abnormality is apparent at birth, but where the inequality of leg length is slight, the diagnosis may not be made until the child begins to walk. To ordinary clinical testing the abnormality is confined to the skeleton; there is no evidence of an associated muscular or nervous abnormality, although in the presence of gross defects muscle attachments may be bizarre. Occasionally there is also a partial or complete absence of the fibula, usually with an associated foot defect. In the present series this has always involved the same side as the femoral defect.

Simple Femoral Hypoplasia (19 cases). The affected thigh is shorter than its fellow, but may appear more bulky. It is often bowed laterally, and the patella is high and small. The leg is held in external rotation, and the hip cannot be rotated internally beyond the neutral position. Radiographs of the femur show no abnormality apart from the shortening, but there is usually some thickening of the cortex of the femur at the centre, and a small beak on its outer side. The radiographs often resemble those of a healed birth fracture, and many of these cases were initially regarded in this light. The distinction is an important one since the congenital short femur always grows less rapidly than its fellow, and the inequality of leg length therefore tends to progress. In this group the definitive shortening does not usually exceed three inches and may well be less.
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These cases have already been considered in detail elsewhere (Ring, 1959).

Congenital Coxa Vara (three cases). Abnormalities in the hips of these patients are rarely detected before they start to walk, although limitation of abduction, and a very slight shortening of the limb, if the lesion is unilateral, are apparent at birth. Where both hips are affected there is a waddling gait, and a pronounced lumbar lordosis. Both abduction and internal rotation of the hip are limited, but the head of the femur can usually be felt in the acetabulum.

In the early radiographs ossification of the head of the femur is usually delayed. The varus deformity of the femoral neck may be apparent soon after birth, but more commonly develops during the second year of life. Ossification defects in the lower part of the femoral neck are common, but not invariable. In this small group mineralization of the head and neck has proceeded relatively normally (Fig. 1). The length of the femur is probably affected mainly by the deformity of the upper end, and the shortening of the limb rarely exceeds one inch (Table). Indeed, true growth in length of the femur, from trochanter to femoral condyles, often appears unaffected.

Femoral Hypoplasia with Coxa Vara (five cases). This type of defect combines the features of the first two groups. The shortening of the limb is usually noticed at birth, or during the early months of life. There is an asymmetry of the thigh creases, and the leg is held in external rotation (Fig. 2). Internal rotation of the hip is absent, and abduction limited. The greater trochanter is elevated, but there is no

### Table

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age (years)</th>
<th>Shortening (in.)</th>
<th>Treatment</th>
<th>Associated Defects</th>
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<td><strong>Congenital short femur:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
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<td>2</td>
<td>1</td>
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<td></td>
</tr>
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<td>2</td>
<td>7</td>
<td>None</td>
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<td>15</td>
<td>1</td>
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<td></td>
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<td></td>
</tr>
<tr>
<td>4</td>
<td>10</td>
<td>4</td>
<td>Stapled</td>
<td>No</td>
</tr>
<tr>
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<td>5</td>
<td>4 (femur)</td>
<td>Extension prosthesis</td>
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<td>7</td>
<td>2 (tibia)</td>
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<td>16</td>
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<td>Amputation through knee</td>
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<td>12</td>
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<td>7</td>
<td>Patten</td>
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<td>10</td>
<td>4</td>
<td>None</td>
<td>Symes' amputation</td>
<td>Partial absence of fibula</td>
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<td>6</td>
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<td>Arthrodasis of knee; Symes' amputation</td>
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<td>5</td>
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<td>Bone grafting</td>
<td>Genu valgum</td>
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<tr>
<td>16†</td>
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<td>None</td>
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<td>Talipes; absent fibula; congenital amputation of arms</td>
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<td>Absent fibula</td>
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<td>5</td>
<td>High boot</td>
<td>Fixed flexion hip and knee</td>
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<tr>
<td>19†</td>
<td>7</td>
<td>4</td>
<td>Caliper</td>
<td>Genu valgum</td>
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* Excluding simple femoral hypoplasia.  † Bilateral.

Fig. 1.—Congenital coxa vara showing deformity of the head and neck and elevation of greater trochanter.
ARCHIVES OF DISEASE IN CHILDHOOD

Fig. 2.—Congenital short femur and coxa vara. There is external rotation of the limb, the thigh is short and bowed.

Fig. 3.—Coxa vara and short femur. The secondary centre for the head of the femur has not yet appeared.

Fig. 4.—Gross shortening of the right thigh associated with congenital pseudarthrosis of the femur.

evidence of hip instability and the head of the femur can usually be palpated within the acetabulum.

Radiographs of the legs show that the affected femur is short, and bowed laterally, within an area of cortical sclerosis on the outer side. The upper end is expanded, the metaphysis is broad, and the epiphyseal centre for the head is late in appearing (Fig. 3). The acetabulum is often shallow. Careful examination of the relationship of the upper end of the femur to the acetabulum permits the diagnosis of coxa vara and short femur to be made before the upper bony epiphysis has appeared. The presence of this centre makes the diagnosis more certain, and reveals the definitive shape of the femur. Defects of this type may be associated with congenital abnormalities of the other femur, of a greater or lesser extent.

In this type of deformity growth of the limb is profoundly depressed, and shortening varies between three inches and six inches at maturity. Even in the absence of gross congenital abnormalities of the tibia or fibula, the growth of the leg below the knee is usually depressed. An indication of this can often be obtained from the early radiographs which show delay in the appearance of the upper epiphyseal centre of the tibia.

**Congenital Pseudarthrosis of the Femur** (six cases). This term has been used to describe the grosser type of femoral shaft defect. The child presents at birth with a very short thigh, although the development of muscle is often apparently normal (Fig. 4). There is usually a flexion deformity of the hip, and abduction is limited or absent. There is commonly an associated congenital defect of the leg or foot.

The early radiographs show a short and narrow femoral shaft, with delay in appearance of the upper and lower femoral epiphyses. In this condition the centre for the lower end usually develops during the first year, although it may show abnormalities of shape, usually associated with a genu valgum. As the child grows the femur increases in length, but its upper end attains a level above the acetabulum, indicating a progressive coxa vara. The centre for the head of the femur is almost always present, and remains within the acetabulum (Fig. 5).
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Fig. 5.—Radiograph of congenital pseudarthrosis of the femur. The shaft of the femur is displaced upwards, but the head remains in the acetabulum.

Fig. 6.—Congenital pseudarthrosis of the femur before exploration showing the shaft of the femur opposite the acetabulum.

Fig. 7.—(The lower part of the figure contains the end of the marrow cavity and 'metaphysis'.) There is a suggestion of columnar arrangement above the cartilage cells and this appears to be a slowly ossifying area.
Thus, although the shaft of the femur appears to be dislocated in relation to the hip joint, there is no true instability, and the head of the femur and the shaft remain in continuity, but not by bone. The upper end of the shaft of the femur is often capped in the older child by a small plate of bone, which probably represents the centre of ossification for the greater trochanter.

Shortening in these patients is due to a combination of coxa vara and a growth defect. By controlling the varus deformity it might be possible to diminish the progress of limb inequality. One child was subjected to operative exploration (Fig. 6). The upper end of the femur was found to be complete in its cartilaginous form, and no true pseudarthrosis was present. The upper part of the metaphysis ended by splaying into the cartilage of the femoral neck, and sections of this region showed the normal process of endochondral ossification (Fig. 7). The cartilage of the upper femur was divided and a wedge removed to abduct the shaft of the femur, and a fibular graft driven across from the upper part of the femur into the cartilaginous head. After this operation the head of the femur rapidly ossified, although para-articular ossification still limited hip movement (Fig. 8).

Hypoplasia of the Lower Femoral Epiphysis (one case). Most congenital defects of the femur affect the upper end either predominantly or completely, although the delay in the appearance of the secondary centre for the lower end indicates that the disturbance of growth may affect the whole bone. In association with defects of the upper part of the femur abnormalities in shape of the lower femoral epiphysis may also appear, and may contribute to the development of a genu valgum (Fig. 9). Complete absence of the lower femoral epiphysis has been seen in one patient. The condition affected both legs, but there was none of the other stigmata of the congenital short femur. It is more likely that this type of abnormality is related to the multiple congenital abnormalities of the epiphyses, rather than the short femur.

Complete Absence of the Femur (one case). This type of defect is usually detected at birth and is commonly bilateral. The thigh is short, and the femur cannot be palpated. The buttocks are broad, and the musculature of the gluteal region is well developed. There are usually associated deformities of the limb below the knee, and deformities of the upper limb may also be present (Fig. 10).
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Although walking may be a little delayed, even in the presence of a bilateral defect, mobility is quite good, although instability is marked. The radiographs of the limb show that the femur is completely absent, but the tibia fails to articulate with the pelvis by some distance. As the child grows a small ossicle representing the lower end of the femur may appear in the radiographs, emphasizing how resistant the lower end of the bone is to this deformity.

Bilateral Defects (three added cases). There are five patients in this series with a deformity of both femurs. In two of them, one with coxa vara, and one with complete absence of the femur, the defect was symmetrical. In the other three patients there was a profound inequality of limb length. The association of two different types of femoral defect in the same patient confirms the view that these defects are all variants of a common abnormality (Fig. 11).
Discussion

Aetiology. Careful inquiry into the family history of these patients, into the history of the pregnancy and that of the birth has failed to find any evidence of a familial factor in this disorder. There is clearly a close association between other congenital defects of the skeleton, and particularly with congenital absence of the fibula. Occasionally both femora are affected. In the simple coxa vara, however, the defects are usually symmetrical, but in the other type of disturbance the femora tend to be unequally affected. The significance of this observation may not be great, since symmetrical disturbance of femoral growth might well pass unnoticed.

In all congenital defects of the femur there is a tendency for the upper metaphysis to be the most severely affected part of the bone. From the study of the serial radiographs in these children it appears that the primary defect lies in the cartilaginous primordium of the femur. It is suggested that the abnormality of structure of this tissue is such that in the minor aberrations it is resistant only to the process of endochondral ossification. Such a resistance accounts for the delay in the appearance of secondary centres of ossification and also for the broad area of weakness in the upper end of the bone. Under the stress of weight-bearing, or possibly of simple muscle activity, the relationship between the head of the femur and the neck becomes disturbed. The development of coxa vara is thus dependent upon a change in the resistance of the cartilage to stress, whilst the failure of growth lies in a resistance to the normal process of endochondral ossification. In the simple coxa vara resistance to ossification is minimal, and shortening is slight, but in the more severe types of deformity changes in the upper end of the femur, and shortening are closely allied.

The association of the congenital short femur with other types of congenital abnormalities of the skeleton is more in favour of a primary disturbance of the cartilaginous framework than a vascular defect, or an intrauterine fracture. It must be conceded, however, that there is no reason why the upper metaphysis of the femur is so commonly involved in this type of abnormality.

Prognosis. The disability in congenital defects of the femur is largely related to the extent of the inequality of limb length. Fixed flexion in the region of the hip joint, although it increases the apparent shortening of the limb, rarely occasions any other disturbance. The externally rotated position of the limb does not give rise to any functional disability, and there is no evidence that it encourages degenerative changes in the hip.

In the individual child, the outlook depends primarily upon the extent of the femoral defect. The shortening of the limb may be predicted with fair accuracy at any age by relating the length of the two femora. Thus in a child with a femur only half as long as the normal one, the shortening as an adult will be somewhere between seven and nine inches. In the presence of coxa vara without other evidence of the congenital short femur shortening is usually insignificant, and in a simple femoral hypoplasia will probably not exceed three inches. Where the two defects are combined the inequality may be as much as six inches and this will be exceeded in the presence of congenital pseudarthrosis of the femur.

Treatment. The presence of fixed deformity of the hip requires treatment in patients with minor femoral defects to permit a normal gait. In the presence of severe defects it may occasionally be necessary to permit a prosthesis to be fitted. Coxa vara without shortening may require subtrochanteric femoral osteotomy, and the results of this operation are usually good (Le Mesurier, 1948).

Relatively few of the children in this series have been subjected to treatment for the limb inequality. Many of them, of course, have not yet reached the age at which definitive treatment is possible. Where the shortening amounts to three inches or less, it is clearly amenable to treatment by stapling the knee epiphyses of the longer limb, or by femoral shortening. The number of patients treated in this way is too few to indicate the value of each of these procedures.

The more severe disturbances of growth have been treated by the provision of suitable appliances. In the presence of a well-developed muscular system, these patients manage to carry a high boot or a shoe with a patten with ease. The function of children in the extension prosthesis is excellent, but the foot tends to be forced into an uncorrectable equinus, thus prejudicing any later attempt to reduce limb inequality. This appliance is also unsightly in the older patient, and some of these children have been treated by Syme’s amputation at an early stage, thus permitting a less obtrusive limb to be worn. The advantages of this procedure must be weighed against the reluctance of the patient and the parents to sacrifice a normal foot.

In the fitting of an artificial limb in the older patient a knee flexion piece is rarely possible. For this type of patient van Nes (1950) has described a rotation-plasty which may well be of value. By fusing the knee and rotating the leg through 180°
the ankle is available to activate the knee-flexion part of the prosthesis.

Summary

A classification of congenital defects of the femur has been presented, and eight examples of this defect have been grouped.

The clinical and radiological picture in these patients, and the growth of the short limb has been described.

The nature of the growth disturbance in these patients has been discussed.

I am most grateful to Mr. J. S. Batchelor, Sir Denis Browne, Professor J. I. P. James, Mr. G. C. Lloyd-Roberts, Mr. K. I. Nissen and Mr. David Trevor who have permitted me to examine and report upon patients under their care.

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P. A. Ring

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