PROXIMAL FEMORAL FOCAL DEFICIENCY

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Children with congenital focal deficiency of the proximal femur present many problems that are but rarely encountered by the individual surgeon who is thus unable to accumulate a wide experience. This paper reviews the literature and analyses the treatment of twenty-three cases at the Royal Children's Hospital, Melbourne, with the object of producing a rational plan of treatment. The management of instability of the hip, malrotation, inadequate proximal musculature and leg length inequality are separately considered for five grades of deficiency. Milder forms are amenable to subtrochanteric osteotomy to correct varus deformity. Exploration and grafting of the pseudarthrosis is indicated where progressive deformity develops. In the more severe deficiency, conservative management of the proximal bony defect provides a better result with an operation only rarely indicated. The gross leg length inequality may be most successfully overcome by Syme's amputation with subsequent fusion of the knee to create an above-knee amputee with an end-bearing stump allowing ready fitting of a prosthesis.

Proximal femoral focal deficiency with its combination of upper femoral maldevelopment of variable degree and femoral shortening, creates a major problem in management. The frequent association with other developmental anomalies in the same leg or in other limbs increases the difficulties which will be encountered before the infant reaches maturity. As the condition occurs infrequently, it is unusual for one person to accumulate extensive experience of it. In an attempt to improve our management of such children, twenty-three patients seen at the Royal Children's Hospital have been reviewed (Table I). Using this experience together with that of previous authors we suggest a plan for future management of the various anomalies.

EMBRYOLOGY

The developing human embryo first shows evidence of limb buds at the 5 millimetre crown-rump (thirty-two days) stage, with rapid and progressive development thereafter. As the apical mesoderm proliferates the limb is laid down in a proximo-distal fashion to be complete at the 12 millimetre stage ten days later (Hamilton and Mossman 1972). During the fifth week individual muscles begin to differentiate, and after mesenchymal condensation and chondrification, ossification of the bones is observed during the seventh week. Differentiation of the primitive limb bud is self-determined, and transplantation at this stage before nerve ingrowth will be followed by normal development. Gardner (1963) has observed that a decrease in skeletal elements may result from failure of the element to form, from failure of the element to develop and grow after formation, or from fusion of two or more elements.

Orderly progression and development from mesenchymal condensation to cartilage, and through calcified cartilage to bone, may be disturbed by some teratological factor (Duraiswami 1952). Numerous agents including irradiation, anoxia, ischaemia, mechanical or thermal injury, bacterial toxins, viral infection, chemicals and hormones have been postulated as a cause. However, only the drug thalidomide has been shown to be a definite cause. When it was taken four to six weeks after conception—during the period of limb bud formation and differentiation—major limb deformities were produced. This defines the period during which any noxious agent produces its effect.

Elements of the ilium and proximal femur develop from a common cartilaginous anlage, with subsequent cleft formation to create a joint cavity (Laurenson 1965). Although movement is essential for development of a normal joint space and sculpturing of articular cartilage, all components of the adjacent bones are present and are of normal size in the absence of movement (Drachman and Sokoloff 1966). This means that if an acetabulum is seen in radiographs at any time in the first year of life a femoral head and neck will be present also, even if not evident in the radiograph. King (1969) emphasised this point after a radiological review of over 100 cases of proximal femoral focal deficiency.

CLASSIFICATION

Despite numerous attempts, a classification which encompassed all varieties of development failure of upper femur and adjacent pelvis was slow to evolve. The first major attempts by Frantz and O'Rahilly (1961)

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Table I. Royal Children's Hospital Series

| Case number	Age (years)	Sex	Side	Type	Distal anomaly	Associated anomaly	Surgical procedures
1	5	F	Left	3	Partially absent fibula	—	Nil
2	7	M	Left	1	Absent fibula; kyphotic tibia	Right talipes equinovarus	Syme's amputation; revision following increasing tibial kyphosis
3	3	M	Left	3	Absent fibula	Right—absent femur, fibula and lateral two rays of foot, arm absent below elbow. Left—humerus and ulna fused, three-digit hand	Hip explored; no reconstruction
4	22	M	Right	5	—	—	Femoro-pelvic fusion; van Nes rotation-plasty
5	2	F	Left	3	—	—	Pseudarthrosis explored; fusion resulted
6	4	F	Right	1	Absent fibula	Mild hydrocephalus	Syme's amputation
7	9	M	Right	5	—	Right arm absent. Left humerus short, micrognathia	Syme's amputation
8	19	F	Left	5	Bifid fifth toe	Right femur absent. Absent upper limbs	Amputation of bifid toe
9	6	M	Left	3	Absent fibula and fifth ray; tibial kyphosis	Right—leg vestigial; arm absent below elbow	Intramedullary nailing of pseudarthrosis and of the tibia; elongation of calcaneal tendon
10	14	F	Left	3	Hypoplastic tibia and dislocated fibula	Left arm absent. Right hand vestigial	Nil
			Right	5	Absent tibia and dislocated fibula	—	
11	7	F	Right	1	Absent fibula; dislocated knee	Left fibula absent, dislocated knee	Bilateral through-knee amputation
12	12	M	Left	5	Absent fibula and lateral two rays of foot	—	Syme's amputation
13	15	F	Left	1	Absent fibula and lateral two rays of foot	Right supernumerary toe	Syme's amputation
14	22	F	Left	2	—	—	Tibial lengthening
15	6	M	Right	5	—	—	Right arm amputation through humerus
16	9	M	Right	3	—	—	Hip explored; no reconstruction
17	1	M	Left	5	Absent fibula and fifth ray of foot	Absent right femur, fibula and lateral two rays of foot	Nil
18	4	M	Left	3	Absent fibula and lateral two rays of foot	—	Syme's amputation
19	21	M	Right	4	—	—	Nil
20	5	F	Right	2	—	—	Nil
21	10	F	Left	2	—	—	Rodding and grafting of pseudarthrosis; Syme's amputation
22	6	F	Left	4	—	Right femur and tibia absent	Rodding of pseudarthrosis
23	4	F	Left	3	—	—	Subtrochanteric osteotomy and plating—repeated
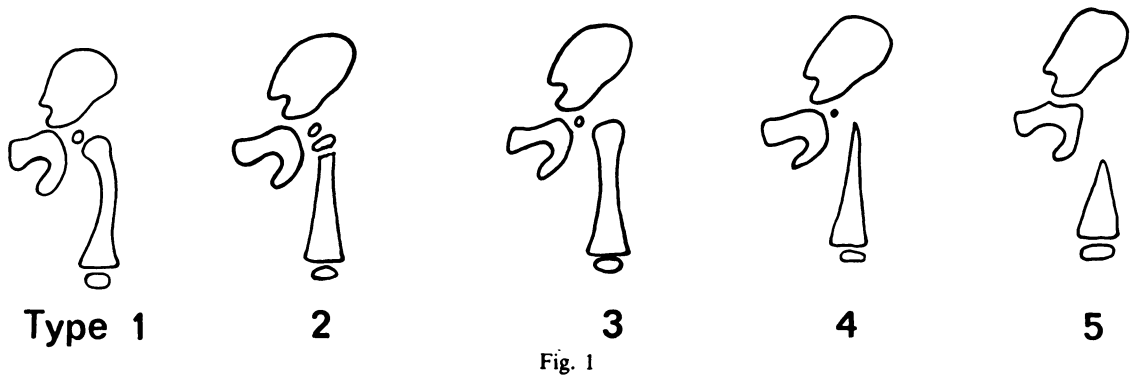
Fig. 1

Fig. 2

Fig. 3

Fig. 4

Fig. 5

Fig. 6

Figs. 1 to 6

Figure 1—Types of proximal femoral focal deficiency. Figure 2—Type 1: short bowed femur with coxa vara; normal acetabulum. Figure 3—Type 2: short femur with subtrochanteric pseudarthrosis and progressive coxa vara; acetabulum normal. Figure 4—Type 3: short femur with bulbous proximal end; acetabulum mildly dysplastic. Figure 5— Type 4: short femur tapering sharply to a point; dysplastic acetabulum. Figure 6— Type 5: short femoral segment; no acetabulum.

and Hall, Brooks and Dennis (1962) did not have terminology adequate to describe a partial deficiency of the proximal femur. More recently Aitken (1969) and Henkel and Willert (1969) have independently proposed classifications which more closely describe the observed deficiency. Henkel and Willert suggested that there was a gradation of deficiency from the mildest, with simple femoral hypoplasia, to the most severe, with femoral aplasia. Aitken first introduced the term proximal femoral focal deficiency to describe partial absence of the proximal femur, and he outlined four groups. However, he omitted the milder form with simple femoral shortening. As the problems in management of the short femur are frequently similar to those where greater deficiency is present, this group should also be included in any classification. Accordingly, the five groups proposed by Amstutz (1969) will be used here (Fig. 1):

Type 1. Congenital short femur with bowing, coxa vara and a normal acetabulum. Although ossification of the femoral capital epiphysis may be delayed, a good hip ultimately results. Sclerosis of the medial femoral cortex with the associated bowing is similar to that observed in milder forms of tibial pseudarthrosis (Fig. 2).

Type 2. Short femur with subtrochanteric pseudarthrosis, progressive coxa vara and a normal acetabulum. Ossification of the pseudarthrosis will follow in most cases but there may be significant residual coxa vara (Fig. 3).

Type 3. Short femur with a bulbous proximal end and delayed appearance of the femoral capital epiphysis. The acetabulum is present and mildly dysplastic. There is a tendency for the pseudarthrosis to ossify but less readily than for Type 2, and the resultant varus may be extreme (Fig. 4).

Type 4. Short femoral segment tapering sharply to a point at the proximal end. There is progressive proximal migration of the tapered sclerotic femoral shaft, with little evidence of spontaneous ossification. The femoral capital epiphysis will ultimately appear, but is often delayed for several years and seldom develops fully. The acetabulum is present and becomes progressively more dysplastic (Fig. 5).

Type 5. Small bony segment representing the distal femoral shaft, with no evidence of the proximal femoral components and no acetabulum. There is a tendency for the femoral segment to elongate but a hip joint does not develop (Fig. 6).

Arthrography may be used to demonstrate a femoral head that cannot otherwise be seen radiologically, but this is largely unnecessary with the knowledge of the close association between the development of the acetabulum and the femoral head. If an acetabulum is obviously present one may safely assume that a cartilaginous femoral head is also present and will eventually be revealed when ossification occurs.

THE PRESENT SERIES

The total number of patients was twenty-three (twelve girls, eleven boys), with twenty-four hips involved, one case being bilateral. The left hip was affected fifteen times and the right nine times. In four cases the deficiency was of Type 1, in three cases it was of Type 2, in eight cases of Type 3, in two cases of Type 4, and in seven cases of Type 5. There was a distal deficiency associated with twelve of the twenty-four hips and other congenital anomalies were present in twelve patients.

MANAGEMENT

Proximal femoral focal deficiency poses four basic problems in management, namely instability of the hip, malrotation, inadequate proximal musculature, and inequality of leg length. These problems will be discussed in turn.

Instability of hip

Instability is of variable severity and related to the extent of ilio-femoral maldevelopment. However, even for Type 1 focal deficiency with femoral shortening and coxa vara a lurching type of gait may be anticipated. This will be magnified in the other types, with persisting pseudarthrosis or failure of joint formation. Although many authors in the past have advocated that the hip be left to develop as it will (Amstutz and Wilson 1962; Bevan-Thomas and Miller 1967; Westin and Gunderson 1969; Meyer, Friddle and Pratt 1971), others have demonstrated that surgical reconstruction has a place (Lloyd-Roberts and Stone 1963; Amstutz 1969). Numerous procedures have been attempted in the past, with variable success. The precise indications for and the timing of such intervention have not yet been determined, but certain points may be made.

Type 1 or 2. The coxa vara deformity can be improved by subtrochanteric valgus osteotomy (Amstutz 1969; King 1969; Westin and Gunderson 1969; Meyer *et al.* 1971). In our series this has not been undertaken as a routine procedure. However, in the other series little technical difficulty has been experienced and union has occurred readily.

Type 3. With a pseudarthrosis of variable extent these cases are more difficult to manage. In our series, seven of the pseudarthroses were explored and cartilaginous continuity between the proximal femoral segment and shaft was confirmed. After excision of the cartilaginous element and fixation of the bony elements in contact by means of a small rod or nail-and-plate, union occurred in four cases and re-exploration was required on one occasion. Westin (1976) advised that the cartilaginous element should be excised to provide bone-to-bone contact if union was to be achieved; however, Lloyd-Roberts and Stone successfully achieved union in two cases by a bone graft across the cartilage; we have had a

similar experience in one case. In general, while the relationship of the femoral shaft and acetabulum is maintained nothing is lost by awaiting further ossification. When proximal migration of the distal femoral segment occurs in Type 2 or 3, exploration with excision and grafting of the pseudarthrosis is indicated. King (1969) believed that bone formation at the pseudarthrosis was stimulated mechanically when the leg was converted to a single lever by arthrodesis of the knee.

Type 4. If these are followed to maturity a hypoplastic femoral head is revealed and this is widely separated from a tapered sclerotic proximal femoral shaft. Surgical exploration in our experience, like that of others, has been unrewarding, and even if union could be achieved the functional result would be poor because of the associated bony and muscular hypoplasia.

Type 5. The absent acetabulum offers little prospect for reconstruction. Alternatives to femoral reconstruction have included: arthrodesis of the distal femur to the pelvis, converting the knee to function as a hip (Doig 1970); excision of the femoral head and neck with transplantation of the fibula (Amstutz 1969); excision of the proximal segment and placement of the femoral shaft in the acetabulum (Westin and Gunderson 1969); various arthroplasties involving acetabular reconstruction and soft tissue interposition; excision of the femur and placement of the tibia in the acetabulum (Cristini 1973); ignoring the bony defect and encouraging stability through increased muscular development resulting from active weight-bearing.

Of these alternatives the last appears to be preferable in most cases, permitting strong movement within a useful range, albeit with some instability. Femoro-pelvic arthrodesis may have a place where gross instability exists, and the resulting proximal migration of the femoral segment threatens to penetrate the skin. While stability is obtained movement will be lost. The one case in this series had only flexion-extension, and although the patient was pleased his gait was not particularly good.

Malrotation and inadequate proximal musculature

These problems are interrelated and will be considered together. Examination of patients with proximal femoral focal deficiencies shows that most have a characteristic posture with flexion, lateral rotation and sometimes abduction deformity at the hip. In this series there was usually a fixed flexion deformity of some 30 to 40 degrees, with 45 degrees or more of fixed lateral rotation. However, the range of movement (often greater than 90 degrees of active flexion and 30 degrees of abduction) was functionally very useful.

No attempt has previously been made to explain this almost characteristic deformity. Examination of the children in our series demonstrated the presence of good gluteal muscles in most while the quadriceps was frequently hypoplastic with a correspondingly small or vestigial patella. Active extension of the knee and flexion of the hip frequently demonstrated a bulky, strongly contracting sartorius. Its action is to draw the leg into the "sitting tailors" position (Last 1966) and this may account for the deformity. Action of the hamstrings and iliopsoas in the presence of a pseudarthrosis of the upper femoral shaft may also play a part.

Despite the deformity, function is good and surgical treatment is not required. Conversion of the leg to a single lever by fusion of the knee results in reduction in the fixed flexion deformity (Westin 1976).

Leg length inequality

As well as the proximal femoral pseudarthrosis and the problems of instability, leg length inequality produces a further major disability. All the femora show hypoplasia and shortening of a variable degree ranging from mild for some Type 1 to gross in Type 5. In general the femur appears to be approximately 20 to 40 per cent the length of the normal. Amstutz (1969), in a long-term follow-up of fifty-seven patients with sixty-eight focal deficiencies, found that the proportional difference in length of the femur remained constant during growth (or increased as proximal migration occurred at the pseudarthrosis), thus confirming the observation of Ring (1959). A few patients with a small inequality in length may be managed by leg-lengthening procedures, or by epiphysiodeses of the normal leg. In this series 50 per cent were associated with significant distal abnormality, usually absence of the fibula and of some elements of the foot. Similar findings have been reported by other authors (69 per cent by Aitken 1959; 50 per cent by Bevan-Thomas and Miller 1967; 40 per cent by Amstutz 1969; 50 per cent by Westin and Gunderson 1969). Amstutz (1969) also observed that deficiency of Type 4 or 5 was more frequently associated with fibular hemimelia.

The sum of the effects of femoral shortening, fixed flexion deformity at the hip and mild hypoplasia of the leg will result in the foot of the affected leg being at approximately the same level as the normal knee at maturity. This progressive discrepancy has been handled in a variety of ways.

Patten. In the young child with relatively short limbs the discrepancy is not so apparent and a suitable patten allows good function (Fig. 7). However, with growth the inequality becomes more obvious and a patten no longer remains functionally or cosmetically acceptable.

Extension prostheses. By placing the foot in full equinus on a platform it is possible to create a prosthesis of appropriate length (Fig. 8). As the discrepancy in thigh to leg ratio increases, this problem may be overcome by placing both the knee and the foot within the socket of an extension prosthesis with a hinge immediately distal

to it (Fig. 9). Although these prostheses improve function they are cosmetically unacceptable (especially to women) and cannot restore a normal ratio of thigh to leg length.

Rotation-plasty. Van Nes (1950) popularised the rotation-plasty procedure previously described by Borggreve (1930). Arthrodesis of the knee and rotation of the distal half of the limb through 180 degrees brought the ankle into a position where it functioned as a knee with the calf muscles acting as the quadriceps. Only one procedure of this nature has been performed at the Royal Children's Hospital (Fig. 10) and the result obtained was similar to those of other series (Amstutz 1969; Westin and Gunderson 1969). Gradual derotation occurred with growth and caused difficulty in fitting a functional prosthesis. Hall and Bochmann (1969) nevertheless apparently achieved satisfactory results in a series of six cases. The development of improved prostheses for the above-knee amputee makes the creation of an artificial knee less important. There are also inherent anatomical difficulties requiring repeated operations and giving a poor cosmetic result. There are currently few indications for the rotation-plasty.

can no longer reach the ground. Even with a discrepancy of three to four inches a child can still play on the beach and swim without difficulty. Although a prosthesis may be fitted easily the marked inequality of thigh and leg length persists.

Arthrodesis of the knee. This has not been performed on any patient in the present series. On examination of these patients the knee is found close to the pelvis with consequent increase in instability of the hip region as the available muscles dissipate their effort across two joints. Fixed flexion deformity of the hip requires flexion of the knee to allow a vertical posture, resulting in an unstable flexed limb. This makes it difficult for the prosthetist to provide a satisfactory socket. King (1966, 1969) has advocated arthrodesis of the knee, giving a single skeletal lever. Such conversion allows the available muscles to operate more effectively across the hip joint, and the limb may then be readily fitted with an end-bearing prosthesis which further stimulates muscle and skeletal development.

At present the management of choice appears to be an early Syme's amputation soon after walking is established with subsequent fitting of an end-bearing

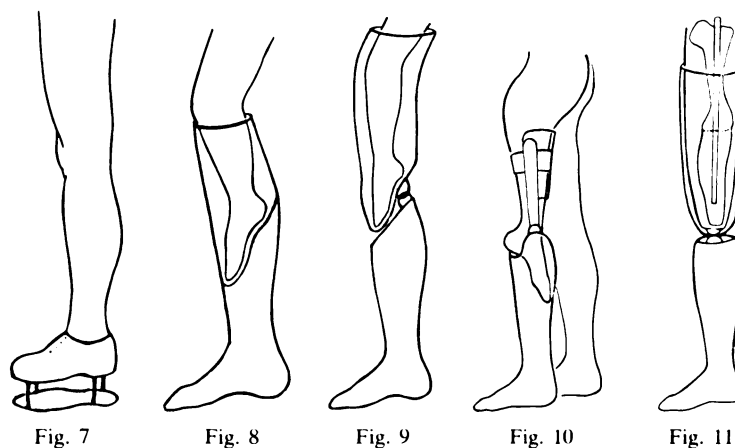Fig. 7   Fig. 8   Fig. 9   Fig. 10   Fig. 11

Figure 7—A pattern of appropriate height to maintain equality of leg length. Figure 8—The foot placed in equinus in a below-knee extension prosthesis. Figure 9—An above-knee prosthesis with the foot in equinus. Figure 10— Rotation-plasty (van Nes) allowing the ankle to function as the knee with an extension prosthesis. Figure 11—Conventional above-knee prosthesis after a Syme's amputation and fusion of the knee.

Syme's amputation. As advocated by Aitken (1959) Syme's amputation has been shown to provide an excellent end-bearing stump allowing ready fitting of a prosthesis. Seven Syme's amputations have been performed in the present series with very satisfactory results. For the large group of patients with abnormalities of the leg and foot this decision can readily be made and accepted by the parents, but where the foot is normal its loss may be more difficult to accept. While procrastination of such a decision should be avoided as the younger child more quickly accepts and adapts to a prosthesis, amputation should be delayed until the foot

prosthesis. With growth and increasing thigh to leg disproportion, arthrodesis of the knee appears to offer considerable advantage. Careful planning should produce a satisfactory lever if appropriate bone is removed and a smooth rod is placed across the joint to ensure continued epiphysial growth. Prediction of the ultimate stump length is difficult because of variable shortening at the pseudarthrosis, femoral hypoplasia and hypoplasia of the leg. Both epiphyses should therefore be preserved initially and a later epiphysiodesis carried out to produce a stump three to four inches above the normal knee at maturity. In this way a mature

limb may be created which is cosmetically and function-
ally comparable to that of other above-knee amputees
(Fig. 11).

DISCUSSION

In writing this paper we have been primarily concerned
with the unilateral cases. The problem is occasionally
bilateral and most authors, including Aitken (1969),
advocate that the disability with its gross dwarfing be
accepted. However, in the presence of normal upper
limbs the above course of management would appear

preferable. Westin (1976) has demonstrated that
bilateral Syme's amputations with above-knee pros-
theses to maintain normal height have been both
functionally and psychologically advantageous. In the
home these children can walk without using their
prostheses.

In the present series one patient with bilateral
proximal femoral focal deficiency also had aplasia of
both upper limbs. For her and others like her the feet are
the only means of independence and their amputation is
absolutely contra-indicated.

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