CONGENITAL APLASIA AND DYSPLASIA OF THE TIBIA WITH INTACT FIBULA

CLASSIFICATION AND MANAGEMENT

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We have reviewed congenital aplasia or dysplasia of the tibia with intact fibula in twenty children (twenty-nine limbs). We have described four morphological groups and four radiological types, and have proposed the clinical and radiological features which in early infancy distinguish these varieties, which vary in their prognosis.

We regard accurate diagnosis to be of the utmost importance in planning future treatment, both in order that unnecessarily proximal amputations or unrewarding operations designed to preserve the foot may be avoided, and that an opportunity to preserve a functioning knee may not be missed.

There are two manifestations of congenital failure of normal development of the tibia. In most cases the tibia is short, and there is associated aplasia or dysplasia of the fibula and reduction of the lateral ray or rays of the foot. Less commonly the tibia is absent or dysplastic whereas the fibula is relatively normal, and the foot may be augmented. A clear distinction must be made between these two varieties of dysplasia (Ollerenshaw 1925; Farmer and Laurin 1960; Henkel and Willert 1969; Hootnick *et al.* 1977).

We are concerned only with the second group, that is, a dysplastic tibia with a relatively normal fibula, and in particular with the recognition of the variations in the anatomy of this anomaly. Our capacity to influence the final outcome is dependent upon our ability to appreciate these variations at an early age.

Apart from a few exceptions, there are three degrees of malformation. First, the tibia is completely absent. Secondly, the proximal epiphysis is preserved with a short segment of tibia. Thirdly, both proximal epiphysis and diaphysis are present but the tibia is short. In all three the fibula is relatively normal in form and development, so that it is readily seen in radiographs in the very young.

Even when tibial components are present, however, their ossification may be delayed, and in consequence the initial radiograph may be misinterpreted as indicating either absence of the tibia when it is in fact merely dysplastic, or a more extensive deficiency than is actually present. Correct interpretation of radiological

Type	Radiological Description	No. of limbs
1	<ul style="list-style-type: none"> • Tibia not seen • Hypoplastic lower femoral epiphysis 	6
	<ul style="list-style-type: none"> • Tibia not seen • Normal lower femoral epiphysis 	12
2	<ul style="list-style-type: none"> • Distal tibia not seen 	5
3	<ul style="list-style-type: none"> • Proximal tibia not seen 	2
4	<ul style="list-style-type: none"> • Diastasis 	4

Fig. 1

Congenital aplasia of the tibia. The four types of deformity as shown radiologically.

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Table 1. Relevant details of twenty cases

Case number	Sex	Side	Radiological type	Foot	Other abnormalities in the same limb	Treatment	Operative findings	Other congenital abnormalities
1	F	R	1a	Normal	None	T-K amputation	Tibia not mentioned	None
		L	1b	Normal	None	T-K amputation	Tibia not mentioned	
2	F	R	2	Normal	None	Upper fibula into tibia		None
3	M	R	2	Normal	None	Upper fibula into tibia		Radial club hand.
		L	2	Normal	None	Upper fibula into tibia		Hypoplastic thumb
4	M	R	1a	Normal	Dysplastic upper femoral epiphysis	T-K amputation	No evidence of tibia	Absent testes
		L	1a	Normal	None	T-K amputation	No evidence of tibia	
5	M	R	1b	Normal	None	T-K amputation	Tibial remnant present. Lower femur well formed	Dextrocardia. Absent testes.
		L	1a	Normal	Femoral spur	T-K amputation	No tibial remnant	Imperforate anus
6	F	R	2	Normal	None	Posteromedial release. Upper fibula into tibia. Lower fibula into talus		None
7	M	R	1b	Normal	None	Treatment refused		None
		L	4	Normal	None	Treatment refused		
8	M	L	1b	Normal	None	Upper fibula into femur. Lower fibula into talus	Tibial remnant excised at operation	Tracheo-oesophageal fistula. Right C.T.E.V.
9	M	L	1b	Normal	None	T-K amputation	Cartilaginous upper tibia-like structure	Right C.T.E.V. Double ureters. Ectopic kidneys
10	M	R	1b	Normal	None	T-K amputation		Haemangioma in loin. Hiatus hernia
11	F	L	1b	Diplopodia	None	Upper fibula into tibia. Syme's amputation	Cartilaginous upper tibia	None
12	F	R	1b	Diplopodia	None	Not known		Imperforate anus. Absent upper limbs
		L	1a	Diplopodia	Proximal femoral deficiency	Not known		
13	F	R	3	Diplopodia	Dysplastic upper femoral epiphysis	Proximal fibula excised. Syme's amputation	Cartilaginous tibia	Abnormal right thumb
		L	3	Diplopodia	None	Syme's amputation	Cartilaginous tibia	
14	M	R	4	Normal	None	None		Imperforate anus.
		L	2	Reduced first metatarsal	None	None		Hypospadias
15	M	R	1a	Normal	Bifid lower femur	None. Patient died		Exomphalos
16	M	R	1b	Diplopodia	None	T-K amputation	Tibia not mentioned	Bilateral augmented radial club hands
		L	1b	Diplopodia	None	T-K amputation	Tibia not mentioned	
17	F	R	1b	Polydactyly	Dysplastic acetabulum	T-K amputation	Tibia not mentioned	Cloaca
18	M	R	1b	Diplopodia	Double fibula	Lower lateral fibula excised. Syme's amputation		None
19	M	L	4	Normal	None	Posteromedial release. Syme's amputation		None
20	M	R	4	Normal	None	Osteotomies of tibia and fibula. Astraglectomy. Both failed. Syme's amputation advised		None

Fig. 2

Fig. 3

Case 4. Figure 2—Photograph showing the clinical features. Figure 3—Radiographic appearances. The features are those of Type 1a.

signs at an early age is essential to rational management. In particular, we must avoid an unnecessarily mutilating amputation through the knee when there is a prospect of preserving the joint in those in whom the tibia, although translucent, is not totally absent.

CLINICAL MATERIAL

We have reviewed twenty-nine affected limbs in twenty patients, nine cases being bilateral (Table I). Our conclusions are based upon the clinical records, operation notes and radiographs of these patients. In addition, we were able to carry out a clinical review of eleven of them.

Fig. 4

Fig. 5

Case 11. Photograph and radiograph showing deformity of Type 1b (compare with Figures 2 and 3). The child has diplopodia.

In our analysis of the structural malformations that are included within this syndrome we are largely dependent upon radiological evidence. We will therefore concentrate upon the interpretation of the radiological signs which are manifest during infancy. Not only is this the age at which most are first seen, but it is also the time when delay in ossification is most likely to mislead the surgeon and perhaps persuade him that the prognosis is so grave that early amputation at a needlessly high level is indicated. Certain clinical features are also important, and these will be described concurrently. Lastly we will consider surgical management.

PRESENTING RADIOLOGICAL FEATURES AND CLINICAL SIGNS

The initial radiograph may reveal one of four types, ranging from translucency throughout the area normally occupied by the tibia, to an appearance which resembles diastasis of the ankle with a complete but shortened tibia (Fig. 1). Furthermore, when there is dysplasia rather than aplasia, new signs will appear as ossification spreads within the cartilaginous anlage, so that one of the presenting alternatives may merge gradually into another less discouraging in appearance.

The tibia cannot be seen (Types 1a and b). This is the most frequent early finding. The tibia may in fact be totally absent (Type 1a, Figs. 2 and 3), or alternatively the proximal part of the tibia may be present, but being as yet an unossified cartilaginous anlage it appears translucent and so misleads the surgeon into concluding that the tibia is totally absent (Type 1b, Figs. 4 and 5). The

Fig. 6

Fig. 7

Case 10. Unilateral, Type 1b, treated by disarticulation. A cartilaginous upper tibia-like structure was noted at operation. The amputation stump is well rounded. The radiograph (Fig. 7) (at age 11) shows that the femur, although of normal configuration, is hypoplastic.

distinction is of paramount importance, for in the latter event we have the opportunity to preserve the knee joint, whereas in the former the alternatives are reconstruction using the fibula to replace the tibia or disarticulation through the knee.

Radiologically the most important distinguishing

feature is the degree to which the lower femoral epiphysis approaches normality. If the femoral epiphysis is translucent in early infancy the proximal part of the tibia is usually absent (Type 1a), whereas normal ossification of the lower femoral epiphysis suggests that the proximal tibial epiphysis is present (Type 1b). The

Fig. 8

Case 4. Radiograph at age 4, showing bilateral deformity of Type 1a treated by disarticulation. Both lower femora are poorly developed and the amputation stumps are conical. There were no upper tibial remnants at operation.

Table II. State of the upper part of the tibia related to the state of the lower femoral epiphysis

	Number of cases	Upper tibial remnant present	Upper tibial remnant absent
Retarded lower femoral epiphysis (Type 1a)	3	0	3
Normal lower femoral epiphysis (Type 1b)	6	6	0

operation notes recorded the state of the upper tibia in nine limbs disarticulated at the knee (Table II). In six of these a tibial remnant was found, and all these limbs had well-formed lower femoral epiphyses. In the other three limbs a tibial remnant was not found, and in these the lower femoral epiphysis was either translucent or markedly retarded. The contrast between the two types of femoral epiphyses persists with further growth, even when disarticulation is performed (Figs. 6 to 10). We believe that if the tibial remnant had not been amputated it would have ossified. Proximal dislocation of the head

Fig. 9

Fig. 10

Case 5. Bilateral deformity of Type 1b on the right and Type 1a on the left. Treated by bilateral disarticulation. On the right side there was a proximal tibial remnant. Note the poorly developed femur on the left (Fig. 10) and the shorter conical stump on this side.

of the fibula is common to both Types 1a and 1b, and does not therefore distinguish between them.

Clinical appraisal of stability of the knee and palpation of a cartilaginous anlage are of limited value. Even if a short tibial segment is present, the knee, lacking the support of the fibula, remains unstable. Furthermore it is difficult both to feel the small anlage as a structure separated from the femur and to decide

whether it is under voluntary muscle control.

The proximal tibia is visible (Type 2). Ossification is already apparent when the child is first seen, and the fibula is dislocated proximally (Fig. 12). However, the morphological state is now evident and the difficulties associated with the Type 1 presentation are resolved. The clinical signs are unaltered unless longitudinal growth simplifies palpation of the rudiment.

Fig. 11

Fig. 12

Figs. 11 and 12

Case 3. Photograph and radiograph showing deformity of Type 2. This child also had a radial club hand.

Fig. 13

Fig. 14

Fig. 15

Figs. 13 to 15

Case 13. Bilateral deformity of Type 3, with diplopodia. Figure 13—Clinical appearance. Figure 14—Radiograph showing absence of proximal tibia. Figure 15—Arthrographs of knees to show well-formed lower femora but poorly defined proximal tibiae.

The distal tibia is visible (Type 3). The distal epiphysis may be visible with mature diaphysal bone development or there may be no more than localised calcified opacity within the tibial anlage (Fig. 14). Proximally, the tibial epiphysis is poorly defined and of uncertain quality. The lower femoral epiphysis is well developed and the upper end of the fibula is dislocated. Clinically the knee is unstable and the foot is augmented. The tibial shaft, whether cartilaginous or bony, is more readily palpable though greatly shortened, and is under voluntary muscle control. Uncertainty about the normality, or even the presence of the proximal tibial epiphysis remains. Only two limbs were available for study. Arthrography demonstrated well-formed lower femora (Fig. 15) but no true knee joints.

The tibia is short, and there is distal tibiofibular diastasis (Type 4). The anatomical situation is clearly shown from the beginning. The upper end of the fibula is dislocated. The foot is normal in structure and is displaced into equinovarus (Figs. 16 and 17). The distal fibula is prominent, extending distally to the lateral border of the foot. This deformity increases as growth proceeds.

ADDITIONAL CONSIDERATIONS

Bilateral involvement. This occurred in nine patients, adding significantly to the final disability. The patient in Case 5 demonstrated total absence of the tibia on one side (Type 1a) and proximal persistence on the other side (Type 1b). We failed to recognise the differences in types and performed disarticulations where remnants persisted. The difference in femoral development continued after disarticulation (Figs. 9 and 10).

Associated abnormalities. Visceral and skeletal deformities in other limbs were very common.

Femur. Apart from the six limbs of Type 1a in which the lower femur was hypoplastic, femoral shortening was uncommon. In contrast, in cases of congenital short tibia with fibular dysplasia, twelve out of fourteen

Fig. 16

Fig. 17

Case 19. Clinical and radiological features of Type 4.

patients have femoral abnormalities also (Hootnick *et al.* 1977). Femoral shortening is an advantage when disarticulation is performed because the prosthetic knee joint may then be properly sited.

Foot. Eight patients had diplopodia (Karchinov 1973), one polydactyly, and in one foot (Case 14) there was a

Fig. 18

Fig. 19

Figs. 18 and 19

Clinical and radiological features of "double fibula". See text.

Fig. 20

Cases 6 and 7. Brother and sister. The boy (left) had bilateral involvement (right Type 1b, left Type 4). His sister had Type 2 deformity on the right.

reduced first metatarsal. There was no relationship between augmentation in the foot and the type of tibial deformity.

"Double fibula". This anomaly is difficult to classify. Two slender bones of fibular configuration do not necessarily represent duplication of the fibula with absence of the tibia. We had one such patient (Case 18), in whom the lower femoral epiphysis was well developed (Figs. 18 and 19). Distally the lateral bone was longer than the medial, displaying the diastasis effect, but paradoxically there was diplopodia (Table I). We are uncertain, therefore, whether to categorise this as a variant of Type 1b or as Type 4. A similar patient was reported by Laurin, Favreau and Labelle (1954) in association with abnormalities of the arms and hands. They regarded the malformation as duplication of the fibula with absence of the tibia.

Aetiology and genetics. There was evidence of thalidomide poisoning in two patients (Cases 3 and 12). In a further three there was a genetic influence. Cases 6 and 7 were siblings (Fig. 20). The father of the patient in Case 16 was similarly affected. In the remainder the cause was unknown.

SURGICAL MANAGEMENT

Treatment should be related to a morphological classification rather than to the presenting radiological or clinical features, useful though they are in distinguishing between the anatomical variants. Morphologically these may be divided into three distinct groups plus one miscellaneous group: total absence of the tibia (morphologically Group I, radiologically Type 1a), persistence of the proximal part of the tibia (morphologically Group II, radiologically Types 1b and 2), and presence of both proximal epiphysis and diaphysis with an overall short tibia (morphologically Group III, radiologically Type 4).

Absent tibia (Group I). It is important to distinguish between aplasia (Group I) and dysplasia (Group II). Our practice has been to explore the knee through an incision suitable for either disarticulation or reconstruction and to proceed according to the findings. Being committed to operation in either event, we have not used preliminary arthrography to resolve the uncertainty. We have no personal experience of Syme's amputation combined with transposition of the fibula to articulate with the femoral condylar notch (Brown 1965; Brown and Pohnert 1972), but cases we have seen do not encourage us to adopt the operation. There is maldevelopment of the femoral condyle and controlling muscles are lacking; so instability and poor voluntary control demand that external support be provided similar to that for an above-knee prosthesis. Moreover, flexion of the knee is often less in the reconstructed joint than in the prosthesis. These factors present greater problems to the limb-fitting surgeon than those posed by

Fig. 21

Fig. 22

Fig. 23

Figures 21 and 22—Case 11. Type 1b. Fibula implanted into the cartilaginous proximal tibia, followed by Syme's amputation. Clinical and radiological result. Figure 23—Case 3. Type 2. Fibula implanted into proximal osseous tibia. Sound union. Parents refused Syme's amputation.

disarticulation, which we therefore believe to be preferable (Parry 1977). In bilateral cases with Types 1a and 1b occurring in the same child (Cases 1 and 5), the predictable inequality of stump length and shape may pose limb-fitting problems.

Proximal part of the tibia present (Group II). The knee is unstable compared with the normal side. The quadriceps and hamstrings are present and inserted correctly. At operation the fibula, which is displaced proximally, is divided at the neck and the distal fragment is opposed to the lateral surface or implanted within the tibial remnant and secured by a Kirschner wire. Although union is seen soon and more convincingly if the tibia is ossified, stability is achieved even when the tibia is cartilaginous (Figs. 21 to 23). Progressive ossification will appear in time. It is preferable to retain the foot initially as an anchor for the plaster, but when union has occurred it is amputated by a modified Syme's technique. There remains some lateral instability of the knee which requires the support of an above-knee prosthesis.

Short tibia with diastasis (Group III). The knee joint is stable and controlled, the fibula is displaced proximally and the foot is anatomically complete but in equinovarus. At the ankle joint, however, the uneven growth between the normal fibula and dysplastic tibia imposes diastasis at the inferior tibiofibular joint and medial inclination, thus causing varus deformity of the foot (Tuli and Varma 1972; Bose 1976). As growth proceeds the deformity increases, and the shortening of the

Fig. 24

Case 19. Type 4. Failed posteromedial release. Fixation of upper fibula to tibia, followed by Syme's amputation. (Mr C. Whalley's case.)

Fig. 25

Fig. 26

Fig. 27

Fig. 28

Figs. 25 to 28

Case 13. Bilateral deformity of Type 3. Figure 25—Operation photograph to show cartilaginous right tibia. Figure 26—The proximal fibula has been excised and a Syme's amputation carried out. Note ossification in the distal tibia. Figures 27 and 28—Clinical appearance two months after operation.

dysplastic tibia is progressive. This shortening will ultimately dominate the problem, so that, even in the unlikely event of its being possible to preserve a plantigrade foot, Syme's amputation with excision of the distal fibular growth plate is clearly indicated (Fig. 24). Various surgical procedures have been carried out in patients of Groups II and III in an attempt to correct equinovarus; all have failed (Table I).

Miscellaneous group. In this group we have included Cases 13 and 18. In Case 13, although there were definite cartilaginous tibiae and well-formed femoral condyles (Fig. 15), the proximal ends of the tibiae were poorly developed. Bilateral Syme's amputations were carried out (Figs. 25 to 28). In Case 18 the lower outer fibula was excised, and thereafter Syme's amputation was performed.

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