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CONGENITAL ABSENCE OF THE TIBIA

By

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Congenital absence of the tibia is a far rarer phenomenon than absence of the fibula (4, 7, 8, 9, 10, 11). But as a result of the thalidomide disaster this malformation, like other defects of the bones of the extremities, has frequently been encountered during the last few years (3, 13).

Since there is considerable difference of opinion concerning the possibilities for treatment of tibial defects (1, 5, 7, 8, 9, 10, 11), we regard it as justifiable to present four cases, involving altogether seven defects of the tibia, treated at the University Children's Hospital and to describe the operative methods employed.

In a previous paper (11) we dealt with the embryological and aetiological aspects and the pathogenesis of this type of defect and shall not, therefore, discuss them in this connexion.

CLINICAL PICTURE

Absence of the tibia may be total or partial. In partial absence of the tibia it is the proximal part that remains (8). In the reports on the few cases of this malformation so far published there has been no mention of the relative frequency of partial and total defects. In total defects the knee joint is also missing and the knee is consequently unstable. In these cases the proximal end of the fibula often lies beside or behind the femoral condyle. In both partial and total absence of the tibia the knee is usually in flexion contracture. The fibula is as a rule comparatively straight (6, 7). On the anterior surface of the leg there is occasionally a dimple (6). In the ankle joint, the foot is as a rule bent into extreme varus position. Defects in the bones and toes on the tibial side of the foot are present in most cases. However, polydactyly may also occasionally occur (8).

The present cases are listed in Table 1. The last of the cases was

Fig. 1 A. *Fig. 1 B.* *Fig. 1 C.* *Fig. 1 D.*

TABLE 1

Case	Sex	Aetiol.	Side	Type of absence	No. of toes	Associated malformations
1. N.L.	male	?	sin	total	5	Pes acquinovarus et diastasis tibiofibularis 1.dx.
2. M.L.	female	?	dx	partial	4	Aplasia radii 1.a
			sin	partial	4	Anomalia costae II sin.
3. M.J.	female	?	dx	total	2	Anus vestibularis
			sin	partial	4 + 1 anomalous	Spina bifida Hernia umbilicalis
4. M.S.	female	Thalidom.	dx	total	6	Phocomelia extr. sup. 1.a Deformatio sacri
			sin	total	7	

caused by thalidomide; in the others the aetiology remains obscure. One case was unilateral, the rest bilateral. Half the defects were total. There were anomalies of the toes in all but one case. Polydactyly occurred in two cases. The femur was within normal limits in most cases. In this respect the defects of the tibia seem to differ clearly from

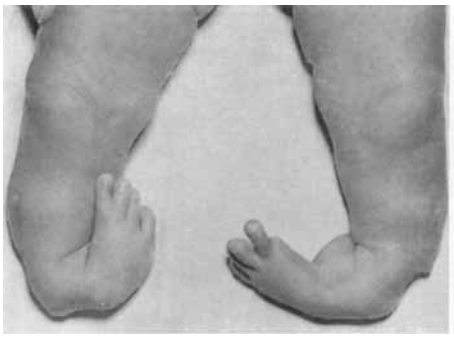

Fig. 2 A.

Fig. 2 B.

Fig. 2 C.

Fig. 2 D.

fibular defects, since the latter are almost always associated with a defective femur (11). As will be seen from the table, other malformations occurred in all cases, the majority of them being severe. This fact, of course, often affects both the planning of the treatment and the prognosis.

TABLE 2

Case	Side	Type	Operations	Age at oper.	Time of follow-up	Result	
1. N.L.	sin	tot.	1. Subperiostal transplantation of right fibula to site of tibia	3 mo		Knee comparat. stable, but ankylosis incomplete	Walks satisfactorily with the aid of a brace
			2. Implantation of distal end of fibula (Fig. 5)	2 yrs 9 mo		Transplants partially resorbed Fibula tibialized, length 50% of contralateral tibia	
			3. Implantation of proximal end of fibula into femur	6 yrs		Good ankylosis in ankle	
			4. Transplantation of metatarsal bone to site of tibia	8 yrs		Marked equinovarus position	
			5. Transposition of proximal end of fibula	9.5 yrs	12 yrs		
2. M. L.	dx	part.	1. Achillotenotomy	6 mo		Knee stable	Walks well with braces
			2. Autotransplantation of rib	10 mo		Fibula fused with tibial shaft and tibialized	
			3. Implantation of distal end of fibula (Fig. 5)	14 mo		Good ankylosis in ankle Persisting equinus position	
			4. Tibio-fibular osteosynthesis (Fig. 6-A)	2 yrs	9.5 yrs	Varus position corrected	
	sin	part.	1. Achillotenotomy	6 mo		Knee stable	
			2. Implantation of distal end of fibula + tibio-fibular osteosynthesis (Figs. 5 and 6-B)	14 mo	9.5 yrs	Whole tibia solid down to the ankle, sligthly bent in medial direction Fibula somewhat atrophied Good ankylosis in ankle, varus position corrected Persist. equinus position	

TABLE 2 (cont.)

Case	Side	Type	Operations	Age at oper.	Time of follow-up	Result	
3. M.J.	dx	tot.	Exarticulation in knee	14 mo	6 yrs	Stump good Configuration and growth of femur normal	Walks excellently with the aid of braces, able to run
	sin	part.	1. Extirpation of supernum. toes	4 mo		Knee stable	
			2. Implantation of distal end of fibula (Fig. 5)	11 mo		Fibula fused to the tibial rudiment and tibialized	
			3. Osteotomy of calcaneus	2.5 yrs		Good ankylosis in ankle	
			4. Tibio-fibular osteosynthesis (Fig. 6-C)	3 yrs	6.5 yrs	Varus position corrected Persist. equinus position	

Fig. 3.

TREATMENT

Without treatment the knee tends to remain in flexion contracture and the foot is fixed in a marked varus position. In total defect of the tibia, in particular, the possibilities of treatment are very limited owing to the instability of the knee. In these cases amputation is generally considered unavoidable (1, 8, 9, 10). In partial absence of the tibia many surgeons amputate, even in early infancy (1). With regard to the age at which amputation should be carried out, opinions vary, but most authors are agreed that it should not be undertaken during the early stages of growth, and there are reports in the literature on the problems to which early amputation has led (1, 12). In some cases efforts have been made to correct the malposition by surgery and to create stability of the knee and ankle by making use of the fibula (4, 8). The late results of these procedures have not been described, however.

The operations performed in the present cases are seen in Table 2. Three patients, and a total of five limbs, were surgically treated. All were presented for treatment when less than 6 months old. In one extremity exarticulation of the knee was carried out. This was a case of extremely severe malformation (Fig. 4–A). In case 2 efforts were at first made to correct the malposition of the foot by achillotenotomy and redression, but with poor results.

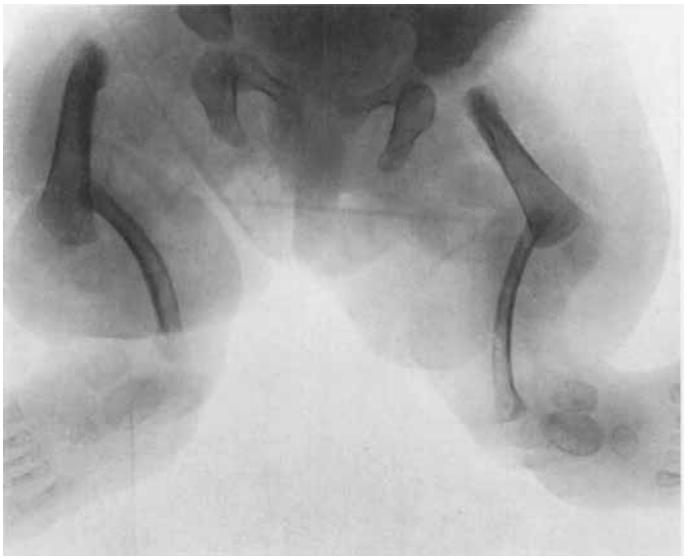

Fig. 4.

In all cases except case 3 (right side) an operation of the type illustrated in Fig. 5 was carried out to correct the varus position and to stabilize the ankle. The distal end of the fibula was exposed and freed from the surrounding tissues. The joint cartilage was excised and the epiphysis made roughly cone-shaped. The upper surface of the talus was exposed and the joint cartilage excised until a smooth surface was obtained. A hole was drilled through the talus to the calcaneus. Taut tibial soft tissue strands were divided sufficiently to allow correction of the varus position of the foot, and the distal end of the fibula was implanted into the drill hole. The fixation was achieved with a Kirschner needle introduced from under the foot through the tarsal bones into the medullar cavity of the fibula. The leg was then immobilized for 6 to 8 weeks in a plaster boot. The needle was removed two weeks after the operation.

In case 3 osteotomy of the left calcaneus was carried out 2 years later to correct the rocking chair-shaped deformity of the foot.

In total absence of the tibia we tried to construct a bony support at the site of the missing tibia by subperiosteal transposition of the contralateral fibula. This was almost entirely resorbed, however. Then we tried to achieve arthrodesis between the fibula and the epiphysis of the femur. Despite several attempts we did not completely succeed.

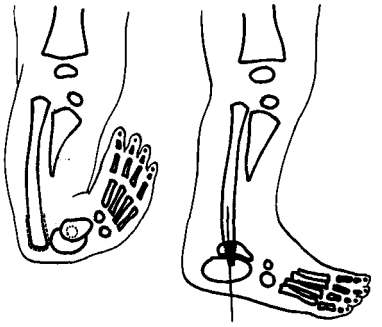

Fig. 5.

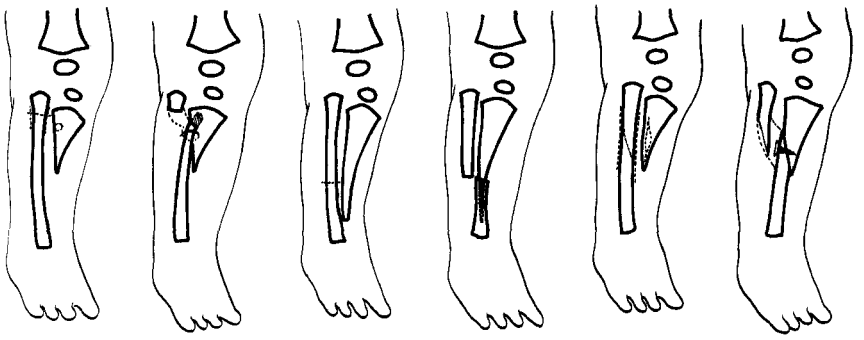

Fig. 6 A. *Fig. 6 B.* *Fig. 6 C.*

In three cases of partial defect we tried to improve the stability of the knee by uniting the fibula with the tibial rudiment. A different technique was applied in each case (Figs. 6 A–C). In case 2 on the right side the cut proximal end of the fibula was implanted in a hole drilled into the side of the tibia and fixed with metal wire (Fig. 6 A). In connexion with the implantation of the distal end of the left fibula in case 2 the fibular diaphysis was cut through and the tibial rudiment implanted into the distal portion of the fibula (Fig. 6 B). Fig 2 D makes a comparison of the end results of these operations possible. In case 3 the proximal end of the left fibula was cut obliquely and united subperiosteally to the side of the exposed tibial rudiment with a screw (Fig. 6 C). Fig. 3 C shows the result of this operation.

RESULTS

Table 2 shows the results obtained. The time of observation varies between 6.5 and 12 years. Exarticulation of the knee-joint carried out in a case of total defect gave a satisfactory result. Despite the normal growth of the femur the stump is well preserved. All other cases were treated by palliative surgery.

In a case of total defect (case 1) the result did not fulfil our expectations. The tibialized fibula constitutes a firm continuation of the femur but the arthrodesis was not quite successful. The pronounced shortening of the tibia renders lengthening by an orthopaedic boot imperative. It is probable that a better walking capacity would be obtained later in this case by above-knee amputation and a suitable prosthesis. The deformity of the other leg also impairs the patient's walking. The marked inward rotation of the foot was later corrected by rotation osteotomy of the leg. The weight-bearing position of the foot then became satisfactory.

In all cases of partial defect the union of the fibula with the tibial rudiment was successful. The knee is stable in every case. The leg is firm and straight and no significant progressive shortening has taken place. The results of all three operative procedures appear more or less equally good.

Good ankylosis of the ankle and correction of the varus position were achieved in all cases. We did not attempt complete correction of the equinus position, which therefore persist in the majority of cases. The foot is in most cases to some extent rocking chair-shaped, since the dorsal end of the calcaneus tends to be displaced in a proximal direction. In case 3 the left calcaneus was osteotomized in order to obtain a better weight-bearing position. The result was excellent.

In all cases walking with the aid of braces is at least satisfactory, considering that there is bilateral deformity of the legs in all of them. Despite a prosthesis on one leg, patient no. 3 is even capable of running.

DISCUSSION

In total defect of the tibia, ultimate amputation seems to be the method of choice, to judge from the present cases. The final result will depend to some extent on the site of the amputation and the age at which this measure is carried out. In practice, the final level of the amputation will be above the knee. If this is done at a very early age,

however, very marked shortening of the stump must be expected. We know from earlier investigations (2) that the distal epiphysis of the femur is responsible for over two-thirds of the growth of this bone. Thus the result may well be the same as in amputation at the middle of the femur. Exarticulation of the knee or palliative operations do not interfere with the normal growth of the femur. In the latter case several operations may be required and a suitable brace must be applied. In these cases the details of the final amputation should be decided with a view to achieving a good stump.

In partial defects, good stability of the knee can always be obtained. A firm and normally growing leg can be achieved by making use of the fibula. In partial absence of the tibia amputation is not to be recommended. A good weight-bearing position of the foot can be achieved by implantation of the fibula in the ankle bones. Even though the equinus position is not completely corrected, it does not appreciably impair walking if an orthopaedic boot is used. After cessation of growth it is possible finally to correct the position, should this be considered necessary.

The tibial defect seems to be bilateral in most cases. This fact naturally renders treatment still more difficult. Case 3, however, shows that the functional result may be quite satisfactory even in very severe cases.

Numerous severe accessory deformities further complicate the problem of treatment. However, recent advances in pediatric surgery have improved the prognosis in many ways.

When the order of importance of the various measures of treatment are contemplated, it must be remembered that to make it possible for the child to move is of paramount importance for its mental and social development.

SUMMARY

On the basis of a series of four patients with a total of seven extremities with a tibial defect, the clinical picture and the methods of treatment of congenital absence of the tibia are discussed. Half the defects were partial. Five limbs were treated operatively. In one case of total absence of the tibia, exarticulation of the knee was carried out at an early age, while another was treated with palliative operations. The authors are of the opinion that in cases of total defect amputation will give the best result but that above-knee amputation should be postponed until growth is complete. Three partial defects were treated by implan-

tation of the distal end of the fibula in the ankle bones and by uniting the fibula with the tibial rudiment. From the point of view of weight-bearing, a good result was obtained in all cases and the patients are capable of walking well with the aid of a brace despite their bilateral defects. The authors do not recommend amputation in these cases.

RESUME

Sur la base d'une série de quatre malades avec un total de sept extrémittés présentant un défaut du tibia, le tableau clinique et les méthodes de tratiment de l'absence congénitale de tibia sont discutées. Dans la moitié des cas, le défaut était partiel. Cinq membres furent opérés. Dans un cas il y avait absence totale du tibia. Une exarticulation du genou a été pratiquée à un âge précoce. Les autres cas ont été traités par des opérations palliatives. Les auteurs sont d'avis que dans les cas de défaut total, l'amputation au-dessus du genou donne le meilleur résultat à condition d'attendre jusqu'au moment où la croissance est achevée. Trois défauts partiels ont été traités par implantations de l'extrémité distale du fémur dans les os de la cheville et par l'union du fémur au rudiment tibial.

Un bon résultat a été eobtenu, par rapport au support du poids, dans tous les cas et les malades sont capables de bien marcher à l'aide d'une attelle, malgré leur défaut bilatéral. Les auteurs ne recommandent pas l'amputation dans ces cas.

ZUSAMMENFASSUNG

Auf Grund einer Reihenfolge von vier Patienten mit insgesamt sieben Gliedmassen mit einem Tibiadefekt werden das klinische Bild und die Behandlungsmethoden des angeborenen Fehlens der Tibia erörtert. Die Hälfte der Defekte war teilweise. Fünf Gliedmassen wurden operativ behandelt. In einem Falle von vollkommen Fehlen der Tibia wurde eine Exartikulation des Knies im frühen Alter ausgeführt, während ein anderer mit palliativen Operationen behandelt wurde. Die Verfasser sind der Ansicht, dass in Fällen von vollständigem Defekt eine Amputation die besten Resultate ergeben wird, dass aber Amputationen oberhalb des Knies bis zum vollendeten Wachstum verschoben werden sollten. Drei teilweise Defekte wurden mittels Implantation des distalen Endes der Fibula in die Knöckelknochen und mittels Vereinigung der Fibula mit dem tibialen Rudiment behandelt. Was die Belastung an-

belangt so wurde in allen Fällen eine gutes Resultat erzielt und die Patienten gehen gut mit der Hilfe von Schienenapparaten, obwohl sie einen doppelseitigen Defekt haben. Die Verfasser empfehlen nicht die Amputation in solchen Fällen.

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