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CONGENITAL ABSENCE OF THE FIBULA

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Examination and management over many years of children with absence of the fibula has made it obvious that the deficiency is not confined to the directly visible anatomical defect, but that it has an affect on the whole leg. The purpose of this report is to submit some observations on children with congenital absence of the fibula who have undergone various forms of treatment.

MATERIAL AND METHODS

In the period 1924 to 1968, 29 cases of congenital absence of the fibula have been treated at the Orthopaedic Hospital in Copenhagen. In 21 cases the absence was unilateral and was not associated with other severe congenital deformities. According to the classification of Coventry & Johnson (1952) all these 21 cases could be classified in groups I and II (partial unilateral absence of the fibula and complete absence of the fibula with minimal or no associated abnormalities).

The distribution according to side and sex is shown in Table 1. In all cases there was shortening of the affected leg, in 18 cases combined with angulation of the lower leg and in 12 cases with a dimple of the skin. A pronounced valgus deformity in the ankle joint was found in 20 cases; in 7 cases there was a valgus deformity in the knee joint probably due to unequal growth of the epiphyses near the knee joint (Figure 1), and in 2 cases the angle of the femoral neck was increased in comparison with the opposite side (Figure 2).

In 9 cases there was a normal number of metatarsal rays in the foot on the affected side, and apart from a slight hypoplasia and a delayed development of the bones, the foot appeared radiologically normal. In the remaining 12 cases there was a decreased number of metatarsal rays with growth abnormalities of the bones in the foot. In 4 cases a bony coalition between the calcaneum and the talus was found.

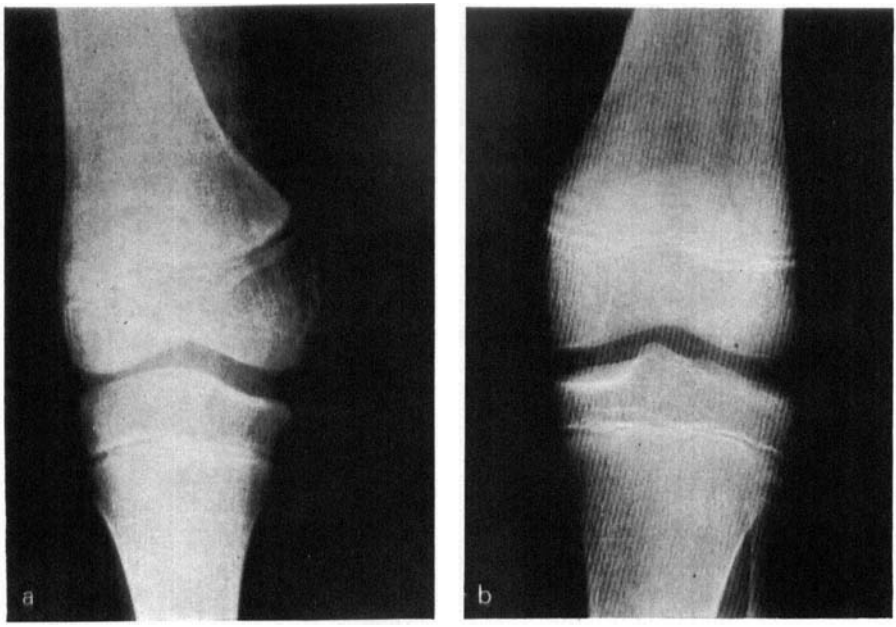
Twelve patients were first seen not later than one month after birth and only 3 patients were over one year of age when first examined. All patients were seen regularly until skeletal maturity was reached or until 1972. From 1952 the normal clinical and radiological examination was supplemented with an orthoradiological examination in order to study the growth rate of the legs. The results of these examinations are shown in Table 2. In the femur the average percentage inhibition

Table 1. The distribution of the 21 cases of congenital absence of the fibula according to sex and side.

	Side		Sex		Total
	male	female	right	left	
Foot with 2 metatarsal rays	1	2	2	1	3
Foot with 3 metatarsal rays	5	–	4	1	5
Foot with 4 metatarsal rays	4	–	2	2	4
Foot with 5 metatarsal rays	4	5	3	6	9
Total	14	7	11	10	21

of growth was found to be 9 and in the tibia 22. The average growth deficiency of the femur was 2 mm per year (0–6 mm) and of the tibia 4 mm (1–9 mm). In 2 patients a femoral length discrepancy of 1½ and 2 cm was found in the first year of life, but later examinations revealed no growth retardation of the femur and the growth rate of the femur on the affected side was found equal to that of the femur on the opposite side.

In order to control the leg length discrepancy, growth stimulating operations and epiphysiodeses were performed. No cases of operative leg lengthening have been done. The stimulating operations were not effective and in only 3 cases could the



Figures 1 a, b. Increased growth of the medial part of the distal femoral epiphysis in a leg with congenital absence of the fibula, in comparison with the opposite leg.

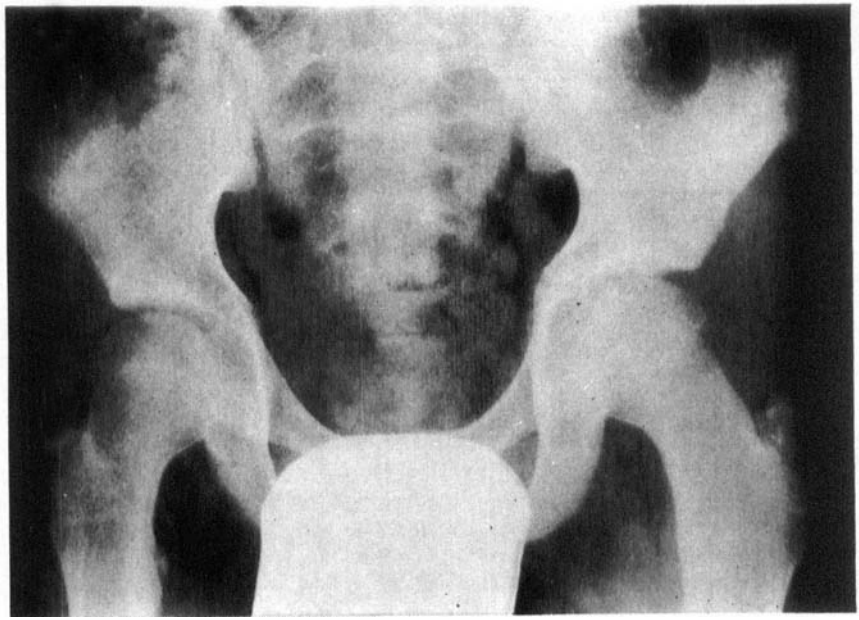


Figure 2. Increased valgus angulation of the femoral neck in a leg with congenital absence of the fibula, in comparison with the opposite side.

results of the epiphysiodeses be evaluated. In one case the estimated correction was obtained whereas only 50 to 75 per cent of the estimated correction was obtained in the remaining cases.

The deformities of the foot were treated with manipulations and corrective casts during the first years of life. In 6 cases resection of the fibular fibrous band was performed. It has not been possible to make on evaluation of this treatment, however.

Out of a total of 8 patients with only 2 or 3 metatarsal rays in the foot on the

Table 2. Results of clinical, radiological and orthoradiological examinations used to study the growth rate of the legs.

		Number of metatarsal rays in foot of affected leg			
		2	3	4	5
The average leg length discrepancy (mm)	Femur	7	20	15	14
as measured in the first year of life	Tibia	60	20	40	17
The average growth retardation (mm)	Femur	3	3½	1	2
per year	Tibia	9	6	4	2½
The percentage inhibition of growth	Femur	5	11	6	10
(average value)	Tibia	35	30	5	10

Table 3. Relationship between foot defect and amputations in 21 cases with unilateral congenital absence of the fibula.

	Total no.	Type of amputation				No. of cases without amputations
		Chopart	Spitzy	Syme	B.K.	
Foot with 2 metatarsal rays	3	1	1	–	–	1 (awaiting amputation)
Foot with 3 metatarsal rays	5	–	1	–	2	2
Foot with 4 metatarsal rays	4	1	–	1	–	2
Foot with 5 metatarsal rays	9	–	–	3	–	6

affected side, 5 have undergone amputation whereas one is awaiting amputation. Four patients presented a foot with only 4 metatarsal rays. Two of these patients have reached skeletal maturity with a leg length discrepancy of 3 and 9 cm respectively, whereas the other 2 patients underwent amputation, one as an adult because of cosmetic reasons and one at 6 years of age because of a leg length discrepancy of 9 cm. Nine patients exhibited a foot, on the affected side, without defects in the metatarsal rays. Three underwent amputation because of leg length discrepancy; in one patient this was mainly caused by a retardation of the growth of the femur. Of the remaining 6 patients in this group 2 have reached skeletal maturity with a leg length discrepancy of 2½ and 3½ cm, whereas the discrepancy in the other 4 younger patients ranges from 4 to 11 cm.

Altogether 10 patients underwent amputation (Table 3). The amputations of the Spitzy-type and through the joint of Chopart were performed in all cases between the ages of 4 and 7 years and were followed by increasing equinus positioning of the hind-foot. In all these 4 cases a lengthening procedure of the Achilles tendon later had to be performed. The B.K.-amputations were performed at the age of 9 years and were followed by re-amputations and later operative revisions. Lastly the Syme's amputations were performed at the ages of 3, 10, 11 and 25 years.

In 3 patients the valgus deformity of the knee gave rise to subluxation of the patella, which later made corrective operations necessary. In all 3 patients amputation had been performed.

DISCUSSION

Congenital absence of the fibula in its characteristic unilateral type was first described by Sachse (1800) (Figure 3). Although the condition is rare an increasing number of cases have been published during the last decade (Farmer & Laurin 1960, Kruger & Talbott 1961, Wood et al. 1965, Mattner 1968, Pappas et al. 1972). As not only the fibula but the whole leg is affected the term paraxial fibular hemimelia as suggested by Frantz & O'Rahilly (1961) is to be preferred. The dysplastic or absent fibula only indicates treatment in cases of valgus deformity in the ankle joint. The associated defects of the femur and the foot, and

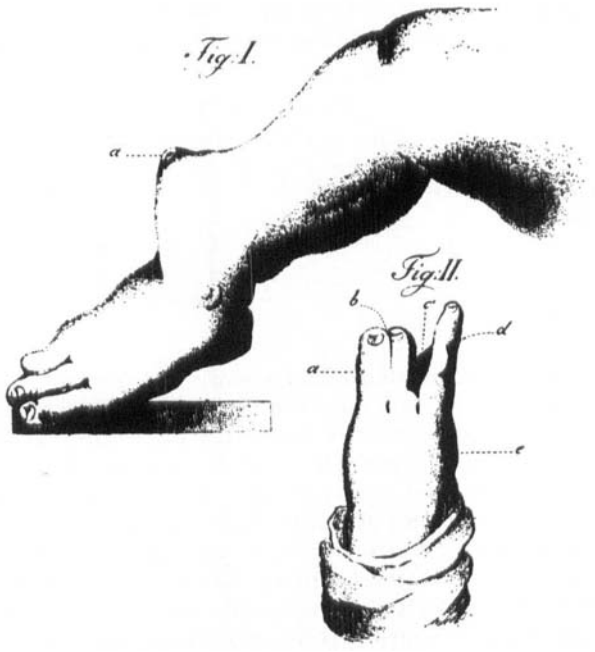


Figure 3. Congenital absence of the fibula. From Sachse (1800).

the decreased growth rate of the long bones in the affected leg are of much greater importance. Thus, for example, the proximal femoral focal deficiency, which often is associated with absence of the fibula is classified by most authors as an independent entity, and only by a few authors (Coventry & Johnson 1952) still looked upon as a case of congenital absence of the fibula.

Congenital absence of the fibula has been classified by Coventry & Johnson (1952) into 3 groups, which were later modified by Kruger & Talbott (1961). In our material this classification was only of limited use. Thus 4 patients who could all be classified in Coventry & Johnson's group I, all exhibited retardation of the growth of the femur with a percentage inhibition of growth between 5 and 10.

In our opinion the problems of treatment in cases of bilateral absence of the fibula and in cases associated with other congenital deformities, differ essentially from the problems in cases of unilateral absence of the fibula. Because of this essential difference, cases of bilateral absence or cases associated with other congenital deformities have not been included in this report.

Two factors are of special importance when treatment is discussed: the abnormalities of the foot on the affected side and the leg length discrepancy. In addition, however, soft tissue abnormalities, associated congenital deformities and age have to be considered. In order to provide and preserve a functional foot, numerous operative treatments have been published (Harmon & Fahey 1937, Serafin 1967) and in order to minimize the leg length discrepancy, growth stimulation, growth inhibition and bone lengthening procedures have been advocated (Pappas et al. 1972).

The aim of the treatment should be to provide the patient with the best possible extremity. In order to obtain this aim, not only bracing, but also multiple surgical procedures may often be necessary. This does not only include osteotomies of the tibia, tendon lenthening, and soft tissue release but also epiphysiodeses on the opposite leg in order to control the leg length discrepancy.

Investigations of the growth rate of the long bones in cases of congenital absence of the fibula seem to be only seldom reported in the literature. Kruger & Talbott (1961) found that the leg length discrepancy was more pronounced in cases with 5 metatarsal rays in the foot than in cases with only 3 rays. This is not in agreement with our findings, which showed an increasing percentage inhibition of growth in the tibia in proportion to the severity of the foot deformity. On the other hand no relationship could be found between the percentage inhibition of growth in the femur on the affected side and the number of metatarsal rays in the foot. In only 2 cases did the percentage inhibition of growth in the femur exceed 10. In contrast to the report of Pappas et al. (1972) the leg length discrepancy as measured at the first examination could not be related to the percentage inhibition of growth of the tibia nor to the ultimate leg length discrepancy.

The treatment has then been associated with multiple surgical procedures and many hospitalizations though the end result has often left a great deal to be desired. In view of these often faulty therapeutic trials and their psychosocial and economical consequences, more authors (Aitken 1959, Kruger & Talbott 1961, Wood et al. 1965) have been advocating early amputation. The indications have been gross abnormalities of the foot and an expected severe ultimate leg length discrepancy.

Amputation, however, might not always be a single definitive operative procedure. In our material more than half of the amputated patients had a secondary operative intervention later. Even if the

problems of overgrowth are put aside, there are still 4 amputated patients with secondary operations. The indication was the progressing valgus deformity in the knee or the equinus position of the foot in patients with amputations through the mid-foot. The rising number of operations on these patients, however, even the amputated ones, may to a certain degree be taken as an indication of a growing interest in the fate of these patients.

CONCLUSIONS

Planning of the treatment must be based upon the defects of the foot and the ankle joint, the ultimate leg length discrepancy and the abnormalities of the soft tissue. If it is decided that a functional foot and ankle joint might be able to be preserved (as estimated from a thorough examination of not only osseous but also muscular and ligamentous conditions) an evaluation of the growth pattern and the percentage retardation of the femur and the tibia should be performed. We think that the early leg length discrepancy is of only limited value in estimating the ultimate discrepancy, and that a careful investigation of the growth rate of the long bones in the affected leg might be of advantage. If it is predicted that the ultimate leg length discrepancy will be under 10 cm, early constructive surgical procedures are indicated.

If a functional foot or ankle joint cannot be preserved, if there is a lack of epiphyseal development, or if the ultimate leg length discrepancy is predicted to exceed 10–15 cm an early amputation might be indicated. In these instances the Syme's amputation probably is the best surgical procedure.

In congenital absence of the fibula virtually no two cases are alike. Even if some general remarks regarding treatment have been made, we want to stress that the treatment here more than anywhere else must be individualized to a high degree.

SUMMARY

On the basis of 21 cases with unilateral congenital absence of the fibula, some observations on the growth rate of the affected leg and the problems of treatment are submitted.

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