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CONGENITAL ABNORMALITIES OF THE FEMUR

A brief review of the cases observed at the Alfred I. du Pont Institute and a report of the treatment of one case of partial absence

By

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The treatment of congenital abnormalities of the long bones, and particularly those of the femur, has always constituted a challenge. This is a report of how such a challenge was met in a child with partial absence of the femur, and a brief report on all of the cases of congenital changes in the femur observed between July 1940 and May 1962 at the Alfred I. du Pont Institute.

The senior author should first like to say how delighted he is to be asked to participate in this homage to two great Swedish physicians who have done so much for the advancement of orthopaedic surgery through their teaching, writings and research, and to wish them long life and many, many more years of productive service.

Congenital abnormalities are constituting an increasing percentage of the patients in the crippled children's hospitals in America as the percentage of patients with poliomyelitis and bone infection is decreasing. At our institution, over a twenty-one year period, the number of patients with congenital malformations has varied from 27.6 to 65.7 per cent of the total admissions, and for four years, from 1953 to 1957, constituted 58.3 per cent of all admissions. The defects and abnormalities of long bones have not been a large percentage of the total number, but have always constituted an interesting and challenging group of cases. In 8500 children examined from July 1940 to May 1962, there were twenty-four who had congenital changes of the femur, representing 2.2 per cent of the approximate 100 major congenital anomalies[1] observed during this period. Twelve of these had a shorte-

[1] Cerebral palsy, congenital developmental errors of the skeleton and hemangiomata are not included.

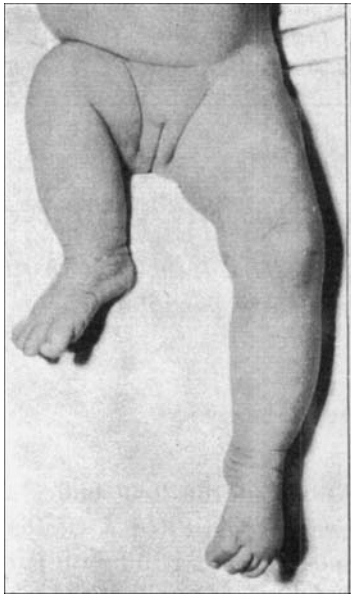

Fig. 1.

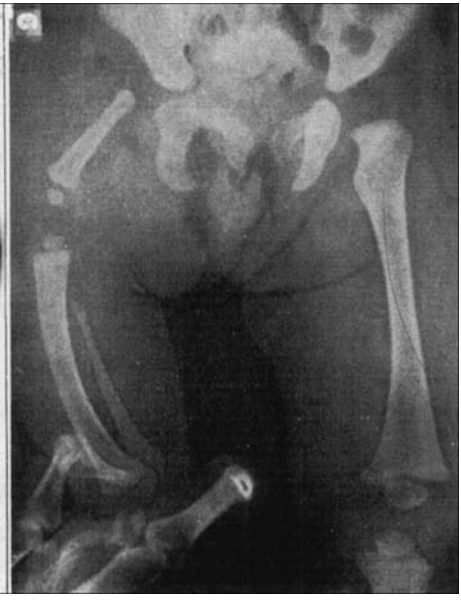

Fig. 2.

Fig. 1. Patient at 5 months of age showing marked shortening of right lower extremity with flexion contracture of hip and knee.

Fig. 2. Roentgenogram at 5 months of age showing partial absence of right femur, lack of development of right acetabulum and absence of upper right fibula.

ning varying from 14.3 cms. to 1.4 cms., and twelve had major defects; seven of the latter were bilateral. Four of the twelve with shortening had an associated absence of the fibula. There were sixteen girls and eight boys. The best classification of the congenital defects of the femur was given by *Max Reiner* in 1901 and is as follows:

I A short femur due to a delayed development with a coxa vara.
II Femur consists of upper and lower ends with one or more bony nodules present in the region of the absent shaft.
III Femur consists of a lower end alone united to the tibia. One or more bony nodules may be present in the region of the upper shaft and head.
IV The upper end of the femur is curved and is some times luxated and not differentiated as to head and neck.
V The hip and knee joints develop but the shaft is greatly shortened.

The nineteen defects in the twelve cases, classified according to *Reiner*, were as follows: 5 in Class I, 6 in Class II, 5 in Class III, 3 in

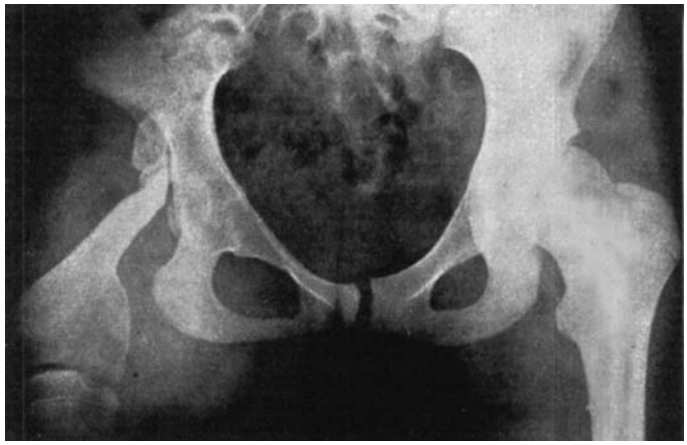

Fig. 3.
Roentgenogram at 12 years 8 months of age showing absence of upper portion of right femur, two bony nodules at upper end of femur and one nodule which probably represents the head of the femur below and medial to upper end of the femur in a small undeveloped acetabulum.

Class IV, and none in Class V. The case reported falls in Class II. Six of the twelve cases had other associated major congenital anomalies. In this latter group there were three with congenital amputations.

CASE REPORT

Girl, RAK, was born March 15, 1946 and first seen when four and one-half months of age. At this time the right lower extremity was 9 cms. shorter than the left. The right hip was flexed and there was a very short thick thigh (Fig. 1). The right foot and lower leg apparently were normal, as were all other parts of the body.

Roentgenograms taken at five months (Fig. 2) showed an absence of the upper two-thirds of the right femur. The acetabulum was shallow. There was a hypoplasia of the right fibula. The right foot was normal. It is interesting to note that the mother had a congenital dislocation of the hip which had had an open reduction when she was four years of age.

At the age of fourteen months an extension brace was applied to the right lower extremity which enabled the patient to ambulate. The child was not seen again at the Institute until October 28, 1958, when she was twelve and one-half years old. At this time she was walking with an ischial weight bearing brace. There was a 90-degree flexion contracture of the right knee with motion from 90 degrees to 150 degrees. The right hip was held in a flexed and abducted position and the thigh was extremely short. Roentgenograms (Fig. 3) showed two bony nodules about the upper end of a thin narrow femur and a third bony nodule below and medial to the upper end of the femur in what looked like a small undeveloped acetabulum. This probably represents what might have developed in to the head of

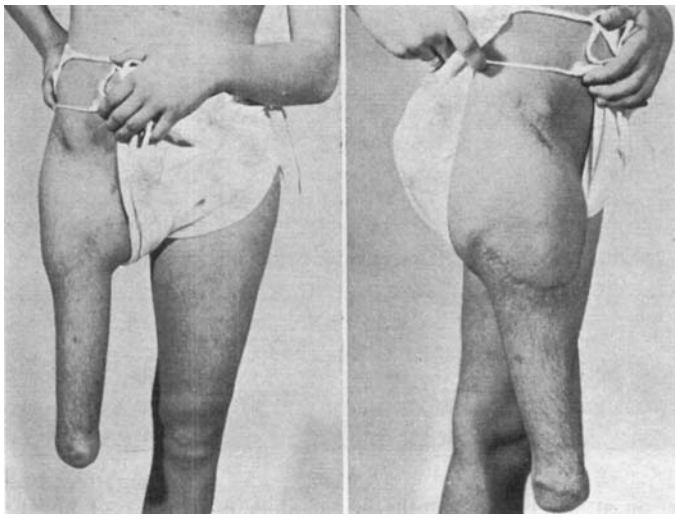

Fig. 4.
Photograph showing right lower extremity amputation stump, front and side views.

the femur. The right knee joint was essentially normal, and the fibula continued to show the hypoplasia.

Since the childe and parents were anxious to have something done which would decrease the unsightliness of the extension brace-prosthesis and enable her to have a better gait, the following was decided upon: The foot would be removed, the knee would be fused, the contracture at the hip joint would be released, and an above knee (AK) prosthesis would be fitted.

When the child was thirteen years old on April 23, 1959, a successful knee arthrodesis was performed and held with Charnley clamps. Two and one-half months later, a soft tissue release of the contracted tissues about the right hip and a Syme's amputation were done. It was not possible to reduce all of the contracture of the hip with surgery. The bilateral hip spica cast which had been applied following operation was twice wedged at the hip on the right in order to decrease the flexion deformity. The lower end of the right extremity was now opposite the level of the left knee joint (Fig. 4). Three months following the last operation the child was fitted with an above-knee quadrilateral socket prosthesis with a pelvic band. (Fig. 5). At the time the prosthesis was fitted the right hip had an extension to 180 degrees from 90 degrees flexion, with a 5-degree flexion contracture; adduction was possible to 60 degrees and abduction to 40 degrees. At first on walking there was a marked gluteus maximus limp which disappeared after gait training.

COMMENT

This case represents a very satisfactory functional and cosmetic result, with which the patient and family were well pleased. She is now

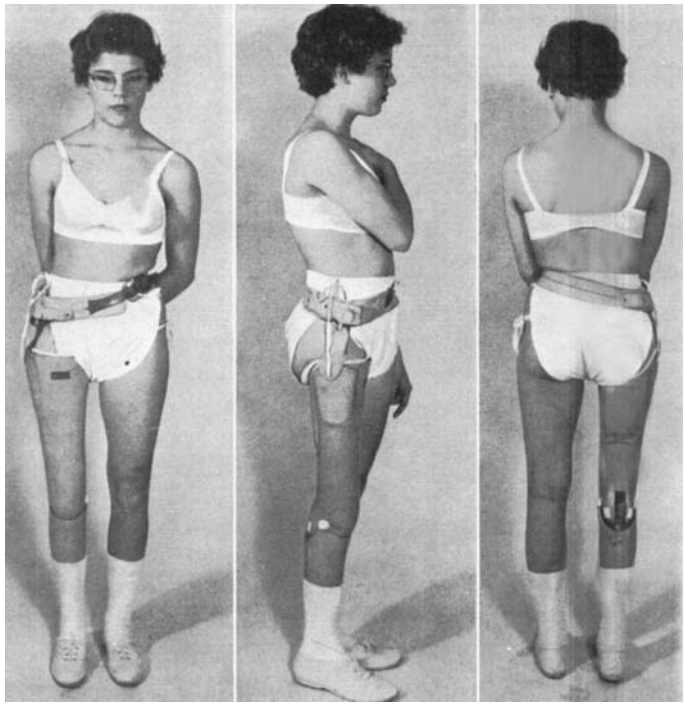

Fig. 5.
Patient at 14 years of age showing right lower extremity prosthesis, front, side and back views.

able to ambulate extremely well and participate in social and school activities without the embarrassment of an unsightly prosthesis.

The procedure employed in this case is not original. *Morris* (7) in 1958 reported a patient for which the same general type of procedure had been done. *Farmer* and *Laurin* (4) had reported previously a case treated in this way. *Morris* has advocated amputation of the foot and a prosthesis for these cases at a very early age. He states that the surgical treatment of these anomalies cannot be standardized because of the wide variety of the type of congenital defects in the lower extremities. The authors certainly concur with this opinion. There are many reports in the literature on the treatment of congenital absence of the femur, but only an occasional mention is made of foot amputation and a prosthesis. In 1959 there was an excellent report of two cases by *M. B. Acker* (1), in which a prosthesis had been fitted without amputation of the foot.

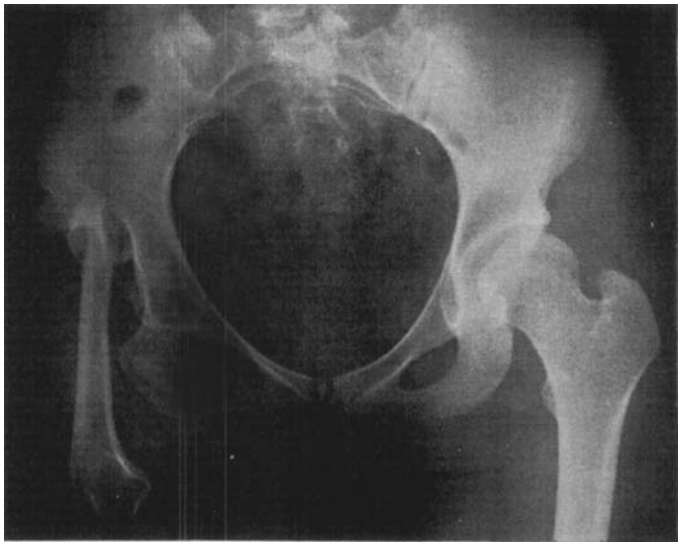

Fig. 6.
Roentgenogram at 16 years of age showing fusion of knee with bony nodules at upper end of femur as seen in Fig. 3.

The rarity of this condition is brought out in a report by *Brussel* (2) in 1939, in which he states that only 104 cases of congenital absence of the femur had been reported up until that time and refers to the work of *Harald Nilsonne* (8) who, in 1928, collected seventy-two cases, ten of which were his own.

One of the best classifications and descriptions of congenital defects of the femur, as well as of the fibula and tibia, was given by *Freund* (5) in 1936, who believed that the short femur was a hypoplasia and represented the mildest form of disturbance of growth; however some authors believe congenital coxa vara to be the mildest form. *Nilsonne* doubts very much whether a general hypoplasia of the femur should be classified with congenital defects. *Ollerenshaw* (9) states that the femur is fifth in order of frequency in congenital defects of long bones; the fibula, tibia, ulna and radius being the first four in this order. There is a very good report of a post-mortem examination of a case by *Manohar* (6) of India in 1939. This contains an excellent description of the muscle abnormalities about the hip joint. The patient was fifty-two years old when he died and had always used a crutch.

One can only surmise as to the cause of congenital anomalies. Some have said that 50 per cent of the congenital malformations are genetic

in character, but others doubt this high percentage and believe it probably to be as low as 20. Since the limb bud for the lower extremity appears in the third week and the first ossification center of the femur appears on the forty-second day, with three centers appearing at the proximal end, one in the middle and one in the lower end, it is quite likely that a disturbance in the development of these ossification centers might be one of the causes. It is the authors' firm belief that most congenital malformations come from a cell nutritional disturbance at the time of the cell division which, in the lower extremity, is from the fourth to the sixth week of foetal life. This opinion is concurred in by *Ollerenshaw.* The excellent work of *Warkany* (13) on vitamins and diet in rats and of *Duraiswami* (3) on hormones in chickens must always be considered when trying to give an explanation of congenital malformations.

SUMMARY

1. A review of the congenital abnormalities in the femur observed at the *Alfred I. du Pont Institute* has been presented.
2. A report on a satisfactory treatment of a case of congenital absence of the proximal end of the femur has been given.
3. A brief review of the literature with a few remarks on the etiology of congenital malformations has been included.

RESUME

1. Présentation d'anomalies congénitales du fémur observées par *l'Institut Alfred I du Pont.*
2. Compte rendu du traitement satisfaisant de cas où il y a absence congénitale de l'extrémité proximale du fémur.
3. Examen succinct de la littérature avec quelques remarques sur l'étiologie des malformations congénitales.

ZUSAMMENFASSUNG

1. Eine Übersicht der angeborenen Abnormiteten des Femurs, die am *Alfred I. du Pont Institut* zur Beobachtung kamen, ist vorgelegt worden.
2. Ein Bericht über die erfolgreiche Behandlung eines Falles von angeborenem Fehlen des proximalen Femurendes wurde gegeben.

3. Eine kurze Übersicht der Literatur mit eineigen Bemerkungen zur Ursache von angeborenen Missbildungen wurde hinzugefügt.

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