CONGENITAL ABSENCE OF FEMUR AND FIBULA

Report of Two Cases

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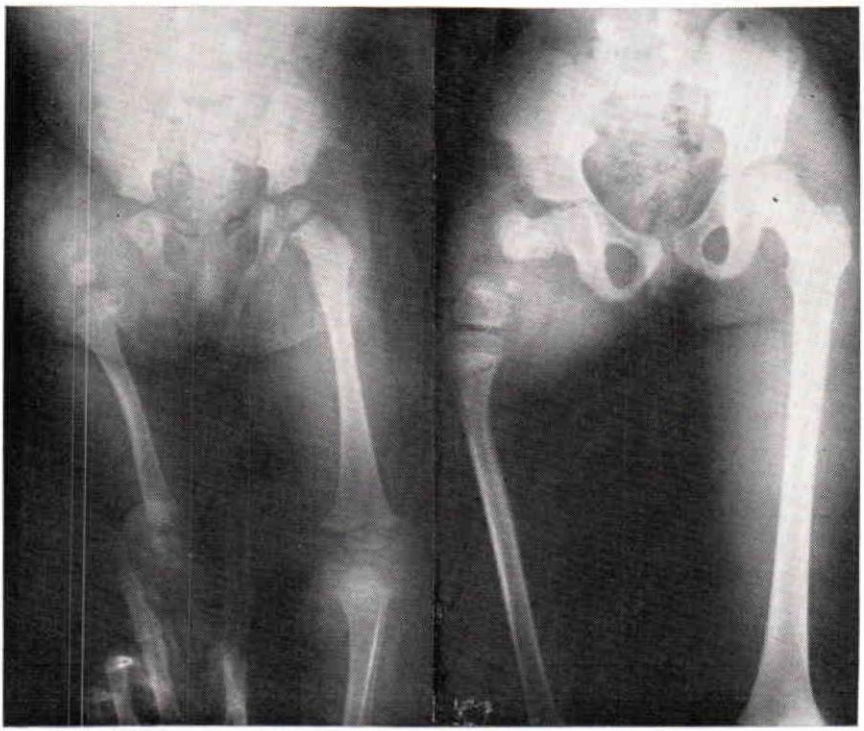
Congenital deformities of the leg are multiform, and almost any pattern may be encountered. Total absence of the femur and the fibula is a pattern that is not common. Two cases of this unusual developmental anomaly are reported here.



Figs. 1 and 2, Case 1. Fig. 1 (Left) Roentgenogram of the pelvis and the legs taken at 1 year of age. The fibula is completely absent, and the tibia is smaller than in the normal leg. The ankle and the foot are normal. A center of ossification, above the tibial epiphysis, is the only representation of the absent femur. The tibia does not form a joint with the pelvis. acetabulum shows fair development. (Right) Roentgenogram at 7 years of age. A rudimentary head and neck of the femur have now appeared, developing from a center of ossification that developed after the initial roentgenogram was made (left). The acetabulum is deep and contains the head of the femur. The initial center of ossification above the epiphysis of the tibia has developed into a bone mass that probably represents the condylar area of the femur. It has no definite structure, and there is no connection between the neck of the femur and this bone mass.

Clinical Description

Case 1. A Negro female child first seen at 2 years of age. An otherwise healthy child with a congenital defect of the right leg. Roentgenographic examination disclosed complete absence of the femur and the fibula, the upper end of the tibia occupying a position in proximity to the acetabulum but not forming a joint. There was indication of a fairly well-formed acetabulum. Later, a center of ossification appeared in the hip area, and, as the child grew and bore weight, this center developed into a recognizable head and neck of the femur, the head articulating with the acetabulum and the acetabulum showing more development. The right leg extended as far as the knee of the opposite leg and was movable in all directions but not with the same range as a normal hip. The leg was reasonably stable, there being some lateral thrust when weight was borne. The foot was of normal contour and functioned well.

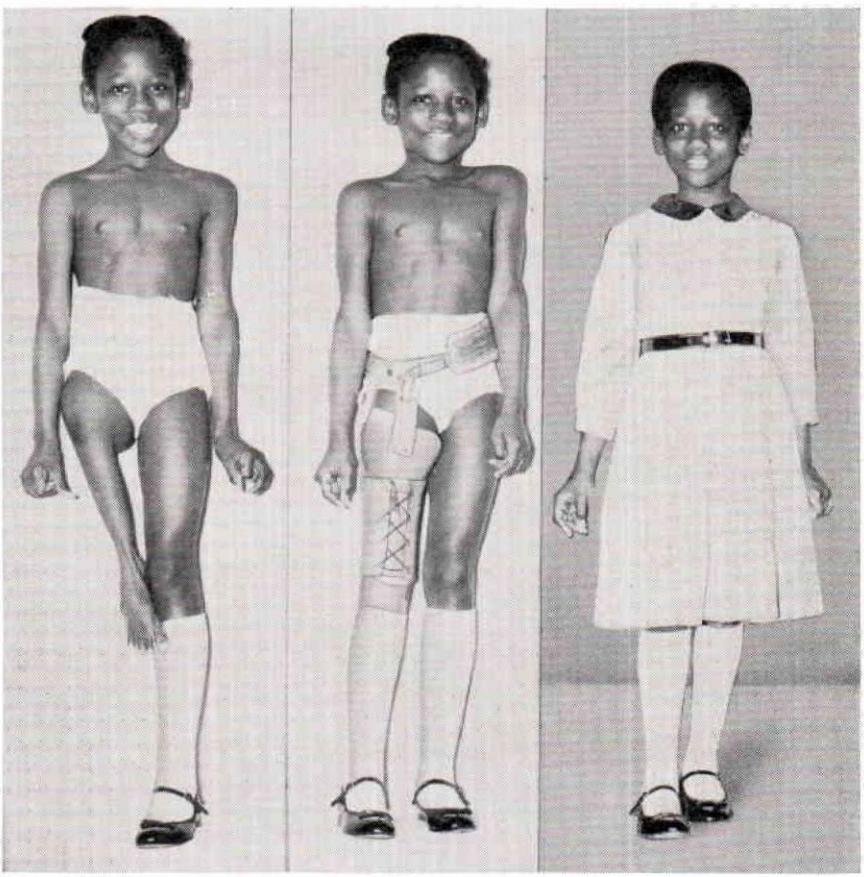
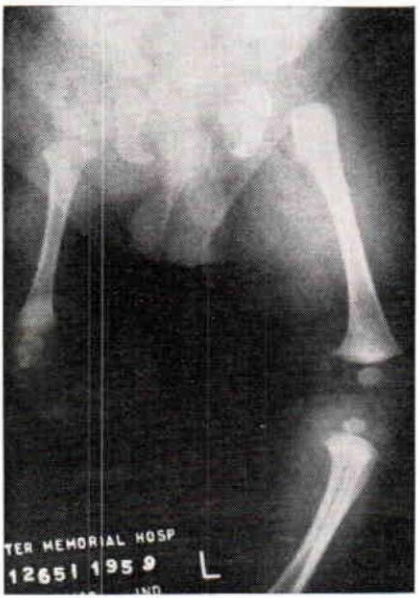


Fig. 2. (Left) Congenital absence of femur and fibula. The deformed leg has a normal foot and ankle joint. It is smaller and less muscular than the lower extremity in the normal leg. Knee and hip joints are absent. Otherwise, the child is well developed and active. (Center) The second type of prosthesis furnished this girl—a well-contoured, nicely fitting apparatus that gives good support. It is tolerated well. (Right) With clothes, the prosthesis is scarcely noticeable. She is a normal, healthy youngster and participates in most activities.

Case 2. A white male child first seen at 3 years of age. An available roentgenogram made when he was 3 months old showed a congenital deformity very similar to that of Case 1. There was a complete absence of the femur and the fibula, the upper end of the tibia being below the acetabulum area and not contacting it. In this case the acetabulum was not developed.

A center of ossification was seen in this early roentgenogram, above the proximal end of the tibia, which center later developed into a segment of bone of no specific contour. There is no direct resemblance of this fragment to femoral condyles or femoral neck, but most likely it represents femoral condyle area. Placement of this bone has a tendency to stabilize the hip area more than before. The foot accompanying this short tibial leg possessed all its components but was a marked clubfoot of the equinovarus type. It became necessary to correct the position by wedging in plaster before the prosthesis could be fitted. The deformed leg was freely movable in all directions, but, as in Case 1, the range of motion was not normal.



Figs. 3 to 5, Case 2. Fig. 3. Roentgenogram made at age of 3 months. The left side appears as the right here. The absence of the femur and the fibula is apparent. The foot is in equinovarus position. The pelvis is very shallow on the deformed side. There is no hip or knee joint.

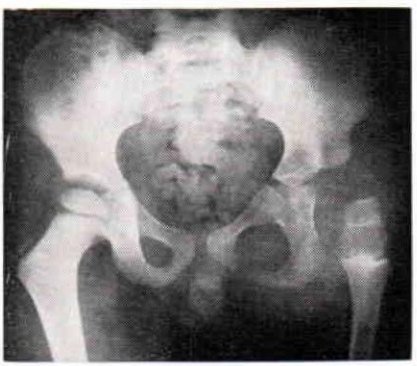


Fig. 4. Roentgenogram at 7 years of age. The left side of the pelvis is not as fully developed as the right side. On the left, the epiphyseal line between the ilium and the pubis and the ischium is open, and the inferior rami of pubis and ischium are not joined. The acetabulum is fairly deep but is not normal in contour. A bone mass has appeared at the upper end of the tibia. It is difficult to evaluate this. It may represent tibial epophysis or condylar area of the femur.

Comments

Before a prosthesis was fitted in these cases, the favorite means of locomotion was by bearing weight on the foot of the short leg and the knee of the normal leg, thus equalizing the weight distribution.

The problem was to supply a suitable prosthesis so designed that sacrifice of any part of the short leg was not necessary, and the foot, a fairly good one, could be used comfortably to assist in weight-bearing and balance. Amputation of the foot, if it seems to be necessary, should be deferred until full skeletal growth has been accomplished. We find the short leg with foot attached to be a very desirable *stump*, free of pressure or

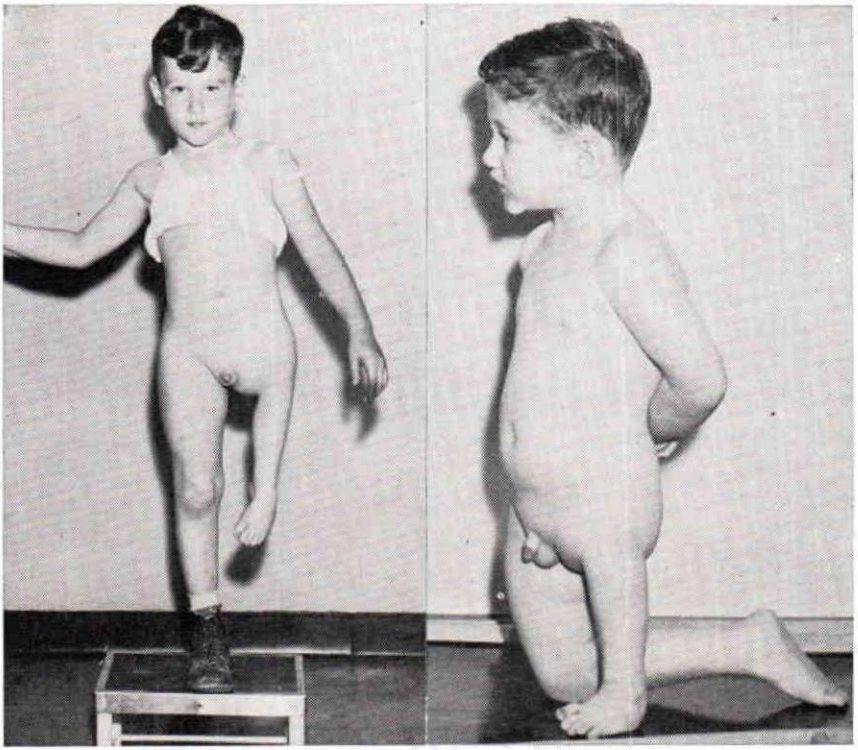


Fig. 5 (Top, left) The relationship of the deformed left leg is well shown here. It is about one half the length of the normal leg as measured from the anterior superior spine to the internal malleolus. Except for the left extremity, the boy was normal physically and quite active. (Top, right) This is the position that this child, as well as the one depicted in Figures 1 and 2, assumed in walking before prostheses were supplied.

abrasion, and comfortable, with weight-bearing at its end. It supplies the prosthesis with more power and easier mobility than a short stump. The chief handicap, with the foot in place, is to devise a workable *knee bend.*

The two main difficulties in function of such a prosthesis are:

1. The problem of a loose *joining* in the hip region, not a true joint; consequently, an unstable point of body support with side thrust.

2. The problem of the foot, which is an important asset in stabilizing and controlling the prosthesis. On the other hand, it is unsightly cosmetically, especially in a female, and adds a difficulty in locating a satisfactory knee-joint bend. However, it is thought that when adult stature is reached, the foot on the short leg will be on the level or above that of the knee of the normal leg, thus solving the proper location of a knee bend in future prosthesis.

The initial prosthesis (Fig. 6, *bottom left*) consisted of a pelvic band—a laced leather cuff enclosing the leg and a shoe for the foot, riveted to a metal plate attached to the side bars. Below this, a contoured artificial leg and foot were attached with a second shoe for weight-bearing. In Case 2, the foot was in equinovarus and was wedged in plaster for better fitting in the shoe. The children learned to walk and balance well with these appliances.

At 6 years of age a better-proportioned, neater and improved fitting prosthesis was supplied: in Case 1, without bend; in Case 2, a bend below the foot. These are now being tested. In both cases the foot was wedged into equinus for better position before applying the last prosthesis.

Summary

Congenital absence of both femur and fibula is a relatively rare condition. Search of the literature disclosed its rarity: references to this particular anomaly were meager. Doctors should be encouraged to report congenital anomalies. Such reports are desirable; they would serve as a valid basis for statistical study.

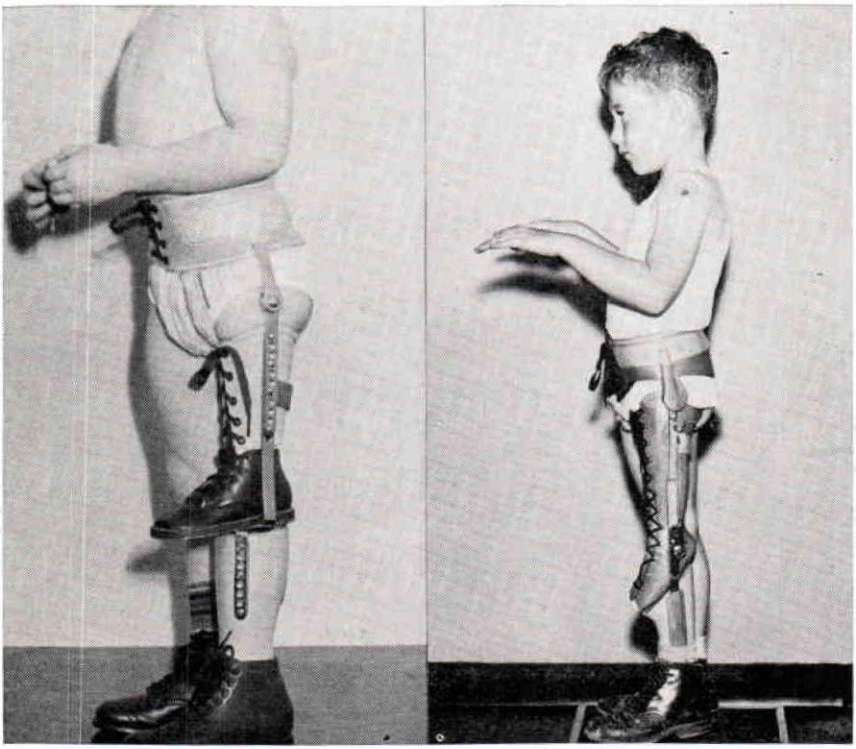
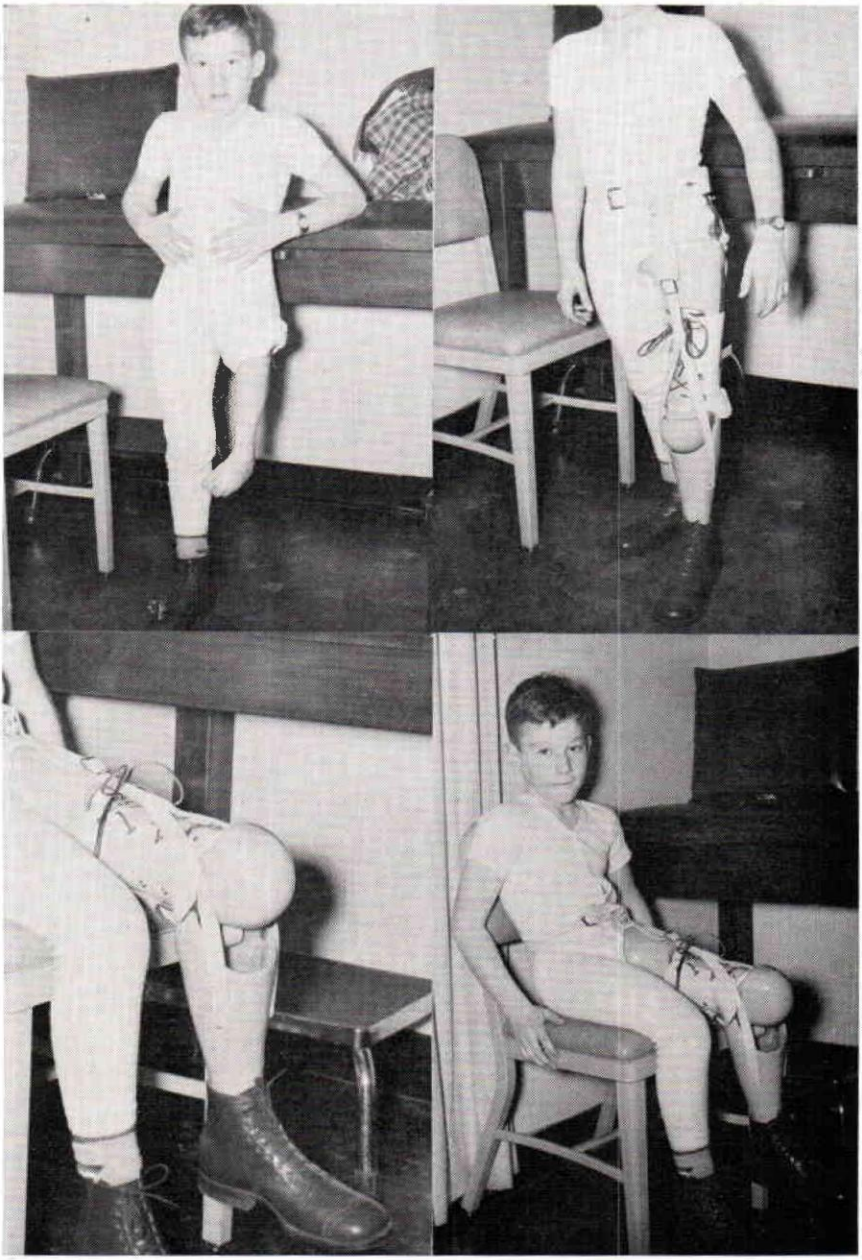


Fig. 6. The initial prosthesis with which Cases 1 and 2 were fitted. They learned to balance and walk quite well with this appliance. (Bottom, right) The second type of prosthesis with which Case 2 was supplied. It differs in construction from the apparatus supplied Case 1. The leg is enclosed in a laced leather boot. The foot has been wedged in equinus and a knee lock put in below the foot with a wire cable release.



EDITOR'S NOTE: The prostheses of these cases were made and fitted by Mr. Walter Pawlowski, C.P., of the Calumet Orthopedic Appliance Company of Gary, Indiana. Mr. Pawlowski is an AOPA member.

The *Journal* is indebted to Mr. Pawlowski for the additional pictures which appear on this page.