Fibular Hemimelia - A Case Report

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ABSTRACT

Fibular Hemimelia (FH) is a rare disorder; it may occur as an isolated anomaly or as a part of a malformation syndrome. We present a case of fibular hemimelia in a 12 year old girl presenting as neglected foot deformity who was treated with external fixator. The brother of the girl had a severer deformity which could not be treated due to lack of willingness on the part of parents. The purpose of the present report is to create the awareness of the condition and to review the literature.

Keywords: Fibular hemimelia, classification, amputations

INTRODUCTION

The incidence of fibular hemimelia has been estimated to be 5.7 to 20 cases per 1 million births1. In most cases, it represents an apparently isolated and sporadic event2. However, FH may be part of a malformation syndrome. Even though this is a rare condition, it is the most common malformation among the long bone deficiency disorders.3

Fibular hemimelia has a clinical spectrum ranging from mild fibular hypoplasia to fibular aplasia. The complete form is more common than the incomplete form; unilateral involvement is more common than bilateral; and the right side is more commonly affected than the left4,5. Bilateral FH is seen in approximately one third of cases. In these cases, the tibiae are often straight. When FH is unilateral, the right side is more commonly affected, and anterior tibial bowing is usually present. In almost all cases, there is deficiency of the lateral foot rays. Less commonly, there is fibular aplasia with a normal number of toes, but almost never is it associated with polydactyly. It is twice common in male fetuses.

Coventry and Johnson6 proposed a classification based upon extent of deformity in associated anomalies, with decreasing prognosis, function, and appearance for each type.

Type I patients have partial, unilateral absence of tine fibula with little or no bowing of the tibia. The extremity is always shortened. There is little or no deformity of the foot. There are no other obvious congenital anomalies in other parts of the body. This type of absence of the fibula frequently escapes diagnosis because of the minimal changes. It may be revealed unexpectedly during roentgenographic examination. Shortening is the most common sign. The degree of absence of the fibula may range from absence of the proximal fibular epiphysis to almost complete absence of the fibula. Little disability is present, and the only treatment required is a heel lift on the affected side or epiphyseal arrest on the normal side.

In Type II tine fibula is completely or almost completely absent. The involvement is unilateral. Tine classical deformity is present, with anterior bowing of the tibia, dimpling of the skin, equinovalgus of the foot, and absence or deformity of the rays and tarsal bones. There is always marked shortening of the extremity. Patients with this type of condition invariably need treatment. The prognosis for function and appearance is relatively poor. Anatomical dissection reveals a fibro-cartilaginous-type band running from the proximal end of the tibia to the calcaneus which is probably the undeveloped fibular analog and a factor contributing to the deformity.

Type III includes bilateral deformities or congenital absence of the fibula of Type I or Type II associated with other congenital deformities. This was the largest group in Coventry's series, involving 55.2 per cent. The condition in most of these cases is difficult to correct, and the prognosis for adequate function is poorer than in patients with Type I or type II deformity.
Frantz and O’Rahilly suggested that anatomical terms be used for all congenital defects to ensure uniform classification\(^7\). Thus using their classification, terminal complete paraxial fibular hemimelia would be used to describe true congenital absence of the fibula and corresponding portion of the foot.

Achterman and Kalamachi\(^8\) proposed a classification system based on clinical as well as radiographic information. In this classification, the congenital deficiencies of the fibula were divided into 2 types. In type I, fibular hypoplasia, included all limbs in which a portion of the fibula was present. This group was subdivided according to the extent of the deficiency. In Type IA, the proximal fibular epiphysis was distal to the level of the tibial growth plate and was often smaller than on the normal side, while the distal fibular growth plate was proximal to the dome of the talus. In Type IB there was partial absence of the fibula: proximally the fibula was absent for 30 to 50 per cent of its length, while distally it was present but did not support the ankle. Type II included all limbs where there was complete absence of the fibula or where only a distal, vestigial fragment was present. According to the Achterman and Kalamachi classification, our case fell into type II.

Authors Experience of two Cases of Fibular Hemimelia:

A 12 year old girl presented to us with neglected congenital talipes equinovarus (CTEV). She was treated with the application of external fixate. Her 8 year younger brother was noticed to have a peculiar deformity of the lower limbs. On examination the boy was found to have bilaterally bowing of the femur (around 90 degrees), absence of fibula bilaterally, absent lateral 2 rays of the foot and deformed hands since birth i.e. fibular Hemimelia type 3. Patients parents were counseled for operative management which included the use of ilizarov fixator and symes amputation, however they were neither willing for complex reconstructive surgery nor for the amputation.

![Fig. 1: a) bilateral acute bowing of the femur with knee flexion contracture b) and c) deformed foot with absent lateral rays and valgus deformity d) deformed hand with absence of a single ray](image1)

![Fig. 2: X-rays showing bilateral bowing of the femur and absence of fibula](image2)
DISCUSSION
Congenital absence of the fibula was first described by Goller in 1896. Fibular hemimelia or absence of the fibula is a rare disorder. The precise etiology of FH is unclear. In congenital absence of the fibula the responsible agents must be active before the eighth week of fetal life, as the limb buds usually appear in the sixth or seventh week. No genetic factor has ever been satisfactorily demonstrated. However, several theories have been suggested, such as defects in the apical ectoderm ridge, defects secondary to an absent anterior tibial artery, and defects in muscle development.

Another proposed theory is that of a disruption of the lower limb developmental field during embryogenesis. The developmental field of the lower extremity includes the pubic portion of the pelvis, proximal femur, patella, anterior cruciate ligament, and lateral or axial foot rays. This developmental field encompasses the commonly associated defects seen with FH, namely, defects of the femur and lateral aspect of the foot. The femur anomalies include congenital femoral shortening, proximal focal femoral deficiency, coxa vara (abnormal angulation of the neck of the femur in relationship to the shaft of the femur), and developmental dysplasia of the hip. The femur anomalies contribute to the overall limb length discrepancy seen in these cases.

Congenital absence of the tibia may be misdiagnosed as congenital absence of the fibula. This is understandable, for the differential diagnosis is sometimes most difficult to make. In gross appearance important points of differentiation are: (a) the foot and ankle in congenital absence of the tibia are in a varus position and no in valgus. The medial malleolus is missing, with resultant medial displacement of the ankle. (b) While atrophy and shortening of the leg are present, as in congenital absence of the fibula, there is little or no anterior bowing of the leg. (c) There is dislocation at the knee. Without the normal articulation of the tibia on tine femur, the fibula rides laterally and there is no true knee joint. The type of deformity of the foot, as mentioned previously, is of aid, and the fact that tine upper end of the fibula is not broadened and does not actually articulate with the femur helps to distinguish it as a fibula. The bowing, if any, is more lateral than anterior when the tibia is missing. The differential diagnosis is therefore extremely important as treatment in most of the cases of congenital absence of fibula is amputation, but this is not the choice in majority of cases of congenital tibial absence. The treating physician is presented with basically a threefold problem: an angulated shortened tibia, foot deformity of an equinovalgus nature, and shortened, contracted soft tissues and in some cases a deformed femur. Prior to 1950 patients were placed on the familiar orthopedic ladder: Cast and manipulation, Hardware, bracing, and shoe build-up. Soft tissue procedures, Bony procedures, tibial osteotomies, tarsal arthrodeses, and malleolar grafts, Epiphyseal arrests and leg-lengthening procedures, as a final resort, amputation.

The intended goals of equalized limb lengths and a suitable foot for weight-bearing were seldom achieved. By 1965 the move to early amputation by Aitken, McCullough, Kruger, Wood and others for the unilateral deformity was complete, becoming the standard of treatment. On the other hand the bilateral case present a treatment dilemma in that amputation is seldom indicated when both fibulae are absent because the two legs in such cases are usually nearly equal in length, usually with functional knees. The indications for early amputation have been stated as 20:

1. Leg-length discrepancy of 7.5 cm (3 in.) or more, actual or predicted at skeletal maturity, or when the affected extremity is or will be 30 per cent or more short than the unaffected extremity utilizing Green-Anderson tables and growth-inhibition factors.

2. Foot deformity when a satisfactory weight-bearing surface is not provided, when conventional shoe wear is impossible and when cosmesis is undesirable.

3. Psychological aspects in our society. The brace is equated to being a cripple, and shortness in stature is an oddity. What other children and parents will accept is unpredictable. A cosmetic prosthetic device arouses curiosity initially, but it is accepted by both patients and peers on a consistent basis. The excellent gait and cosmesis do not select patients out as “different” or as “an oddball.” Family support is maximized by decreased hospitalization time, with an added fourth benefit of streamlined economies in number and length of hospitalizations.

4. Economic considerations: in a developing country like ours, many people are willing to afford the cost of multiple reconstructive surgeries and amputation and prosthesis offer a good treatment option even in bilateral cases.

The question of whether to do a Syme- or Boyd-type amputation preserving the distal tibial epiphysis appears as a surgeon’s choice. The Boyd type provides, in theory, additional length, eliminates potential migration of the fat pad, preserves septa and cushioning effect of the heel pad, and has less chance of disrupting the calcaneal artery, thus resulting in a dead flap. Both procedures prevent bony overgrowth of the tibia; the best results are obtained when the heel pad is aligned in a plantigrade fashion. The Syme technique remains the method of choice when the calcaneus is in marked equinus position.
REFERENCES


