

## FIBULAR HEMIMELIA: A CASE REPORT

\* K.C Eze, \* A.O Akhigbe, \*\*G.O.G Awosanya

\* *Department of Radiology, University of Benin Teaching Hospital, Benin City, Edo State, Nigeria.* \*\**Department of Radiodiagnosis, Lagos University Teaching Hospital, Idi-Araba, Lagos, Nigeria.*

### ABSTRACT

Fibular hemimelia is the congenital absence of the fibula and it is the most common congenital absence of long bone of the extremities. Shortening of the extremity is obvious at birth with leg-length discrepancy. On plain radiograph of the leg and foot, significant fibular deficiency (hypoplasia) or absence of the fibula can be seen. In this case report, a 6-month-old baby boy with fibular hemimelia is presented. The radiological diagnosis and differential diagnosis are discussed. The treatments which include repeated corrective osteotomies and leg-lengthening surgery are costly and associated with residual deformity.

**Key words:** Fibula, Hemimelia, Radiology, Diagnosis

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### INTRODUCTION

Fibular hemimelia is the most common congenital aplasia of a long bone, it is followed by aplasia of the tibia, ulna, radius and femur in that order<sup>1</sup>. Shortening of the extremity is obvious at birth with leg-length discrepancy of up to 19%. Twenty eight percent of the leg length discrepancy is contributed by the tibia and 13% by femur<sup>1-5</sup>. Proximal focal femoral deficiency (PFFD) occurs in 20% of patients with fibular hemimelia while fibular hemimelia occur in 50% of patients with proximal focal femoral deficiency<sup>4</sup>.

### CASE REPORT

Master AO, a 6 month old male baby born to a 28 year old house wife and a 33 year old trader-father presented to the general out patient clinic of University of Benin Teaching Hospital (UBTH) on 4<sup>th</sup> of January 2003 having been referred from a private hospital where he was delivered. The mother complained of shortness of the right leg of the baby from the knee to the foot, lateral curvature of the right foot from the ankle and general unhappiness since she gave birth to the baby. Pregnancy and delivery were uneventful. The baby is the second child of the family in a monogamous setting. The other sibling is a female and healthy. There was no history of congenital anomaly or physical deformity, hypertension, diabetes, asthma, sickle cell disease, drug intake or serious alcohol intake in the family. On examination, the patient was healthy looking, conscious and alert and showed normal development milestone. The weight was 6 kg. Musculoskeletal system examination showed gross shortening of the right leg from the knee to the foot.

There was equinovalgus deformity of the right foot. The right thigh, the left lower limb and other parts of the musculoskeletal system were normal (fig 1). Plain radiograph of both lower limbs and the pelvis showed a short right tibia with medial bowing of the bone at the mid portion (fig 1). The talus was vertical. There was absence of the last two lateral digits of the right foot. The right femur and the left lower limb were normal (fig 1).

Ultrasonography of the right leg showed an echogenic soft tissue extending from the proximal to the distal aspect of the right tibia along the concave border. An impression of fibular hemimelia was made and the patient was referred to the orthopaedic surgeon.

**Figure 1: Plain radiograph of both legs showing absence of fibula of the right leg. The left leg is normal. The tibia of the right leg is curved.**



## DISCUSSION

Fibular hemimelia is a rare disease caused by congenital longitudinal deficiency or absence of fibula and it represents a spectrum of deformities<sup>1,2</sup>. It ranges from mild fibular hypoplasia to aplasia and may be unilateral or bilateral. Fibular hemimelia can be an isolated deformity of the lower limb but often it is associated with proximal focal femoral deficiency (PFFD), abnormalities of the tibia like absent tibia, bowed tibia, deformity of distal tibial epiphysis resulting in convexity with lateral and posterior slope of distal joint surface. This deformity results in equinovalgus position of the foot and ankle and frequently consecutive dislocation of the foot. Tarsal coalition can result. There may be absent phalanges usually on the lateral side (as noted in this case) of the foot. Soft tissue changes are involved in some cases. Up to 80% have anterior bowing of tibia and one third have also medial bowing with sclerosis on the side of concavity. The apex of the tibial convexities is at the junction of middle and distal third. In 60% of the cases a light soft tissue band can be palpated running from the proximal portion of the tibia to the calcaneus and often contains cartilage and may be the remnant or the equivalent of the absent fibula<sup>3-5</sup>.

There are different classifications by different authors<sup>4,6,7</sup> but the most popular on the basis of guide to treatment and functional restoration is that by Kruger<sup>4</sup>.

### Type I. Unilateral involvement

- Ia. Normal femur
- Ib. Presence of proximal focal femoral

deficiency

### Type II. Bilateral defect

- Ia. Normal femur
- Ib. Presence of proximal focal femoral

deficiency<sup>4,5</sup>

On plain films of leg and foot, a significant fibular deficiency (hypoplasia) or absence of fibula can be seen. Tibia is often bowed anteriorly or / and medially with cortical sclerosis along the concavity of the curvature. The epiphysis of tibia may be small or absent due to general delay in appearance of ossification centre in this condition. But generally there is deformity of distal tibia epiphysis. Tibia articulation with femur is normal. There is general delay or absence in appearance of pedal bony structures with ossification occurring as late as 5 years<sup>1,7</sup>. In this patient the talus on the affected side had not appeared and the metatarsals and phalanges of the fifth digit were absent. Various degrees of soft tissue and bony abnormalities of the lateral aspect of the foot have been associated with the syndrome<sup>4</sup>. Plain radiography of the hip is necessary to exclude

proximal focal femoral deficiency. The femur was normal in this case. Several authors have reported antenatal sonographic diagnosis of fibular hemimelia and their findings included non-visualization of fibula and bowed, shortened tibia<sup>8,9</sup>. Several treatment modalities which include correction of the clubfoot and tarsal coalition exist<sup>14,10</sup>. Accurate diagnosis of the lesion and concomitant club foot as well as defective cruciate ligaments of the knee using ultrasound and magnetic resonance imaging (MRI) is necessary before a decision on the best method of treatment is made. This may involve osteotomy or leg lengthening procedures<sup>10-12</sup>.

In long term follow up of four cases in a series by Tomas et al<sup>1</sup> and in other series by Bradish<sup>12</sup> and McCarthy et al<sup>13</sup>, it was discovered that several leg lengthening operations may be necessary and simple amputation may be the treatment of choice in severe cases of fibular hemimelia.

Differential diagnosis of fibular hemimelia on plain X-ray may be difficult but careful inspection of the radiograph will usually lead to diagnosis even if the tibia is small and slender to mistake it for the fibula. In tibial hemimelia, the fibula will not be bowed and does not articulate with the femur. The foot is in varus position in tibial hemimelia whereas it is in valgus position in fibular hemimelia<sup>4</sup>. In anterior or medial congenital bowing, both bones are present and the tibia is well articulated with the femur. Both bones are also present in congenital pseudo-arthritis of the tibia and there is a break in continuity of tibia and the medullary canal is interrupted<sup>4</sup>. In Nigeria where the poverty level is high, the people do not generally accept amputation due to sociocultural reasons. Repeated corrective osteotomies required in this condition are expensive<sup>13</sup> and thus the disease can be an economic tragedy for a family that has one.

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