CASE REPORT

FIBULAR HEMIMELIA: A RARE OCCURRENCE IN KARACHI, PAKISTAN. BILATERAL COMPLETE ABSENCE OF FIBULA ASSOCIATED WITH BILATERAL PROXIMAL FOCAL FEMORAL DEFICIENCY AND CLUB FEET ON THE SKELETAL SURVEY

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ABSTRACT

Fibular hemimelia is a non-lethal congenital lower limb skeletal malformation. It encompasses a broad range from partial to complete absence of fibula. Its incidence is very low worldwide. It affects boys more than girls in a ratio of 2:1. It may occur as an isolated anomaly or associated with other lower extremity anomalies like genu valgum, tibial bowing, ball and socket ankle joint and absent lateral foot ray anomaly. It may be associated with other syndromes including Fuhrmann syndrome, Al-Awadi/Raas-Rothschild syndrome, Roberts syndrome, and femur-fibula-ulna syndrome and FACTO syndrome. This case is not only unique as a rare occurrence in Karachi, Pakistan, but also in bilateral involvement, demonstrated complete absence of fibula i.e. type II fibular hemimelia; in association with a bilateral proximal focal femoral deficiency with pseudoarthrosis and club foot on each side.
Keywords: Fibular hemimelia, skeletal survey, conventional imaging or x-ray, proximal focal femoral deficiency, pseudoarthrosis, club foot.

Introduction

Fibular hemimelia is a partial or complete absence of fibula.[1] The etiology of this malformation is unknown.[2] Its incidence is very low. It occurs in 5.7 to 20 cases per one million births. Less than 1000 people have been reported in the literature in the United States.[3] It affects boys twice as girls. It is associated with other syndromes including the Al-Awadi/Raas-Rothschild syndrome, Roberts syndrome, and the femur-fibula-ulna syndrome and FACTO syndrome.[4] It may be isolated or associated with other associated anomalies of the lower extremity like genu valgum, tibial bowing, ball and socket ankle joint and absent lateral foot ray anomaly.[5]

Here, presenting a case of bilateral fibular hemimelia associated with two unique lower extremity anomalies including proximal focal femoral deficiency and club feet.

Case Report

One and half-months-old boy presented in medical OPD of National Institute of Child Health (NICH) along with parents, having bowing of both lower limbs since birth. It was a product of non-consanguineous marriage and was born via vaginal delivery. His birth weight was 3kg. APGAR score was satisfactory at 1 minute

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and 5 minutes. Antenatal History revealed no rash development, fever, or drug and radiation exposure. On clinical examination, both lower limbs were bowed posteriorly. Bilateral medially oriented fore feet was recorded. (Fig.1). Systemic examination was grossly unremarkable. Neonatal reflexes were appropriate. He was advised for an X-ray pelvis and both lower limbs which showed a normal pelvis, bilateral proximal focal femoral deficiency with pseudoarthrosis and complete absence of fibula. Tibia was also curved as mentioned in (Fig.2). X-rays both revealed parallel organization of talus and calcaneum with forefeet inversion which correspond to clinically visible club feet. The rest of the skeletal survey was unremarkable.

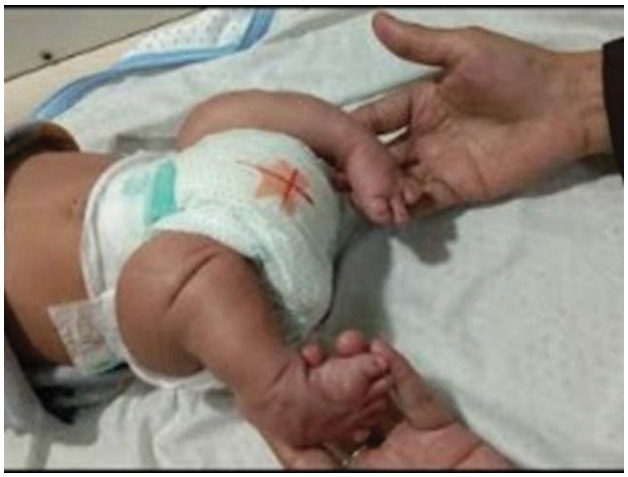
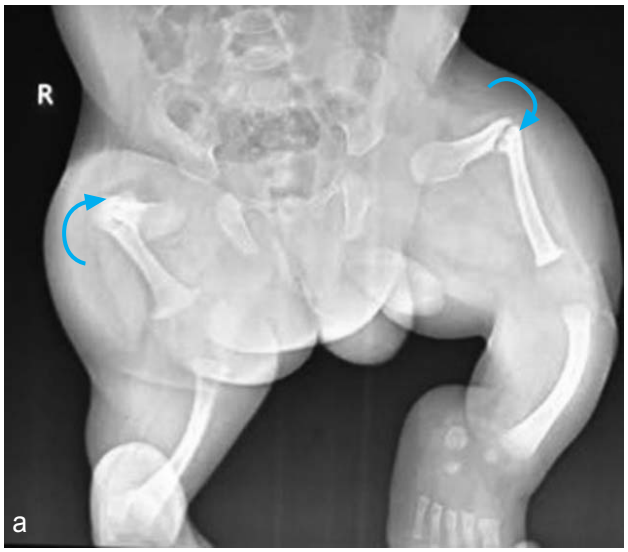


Figure 1: The clinical picture of congenital owing of both lower extremities.



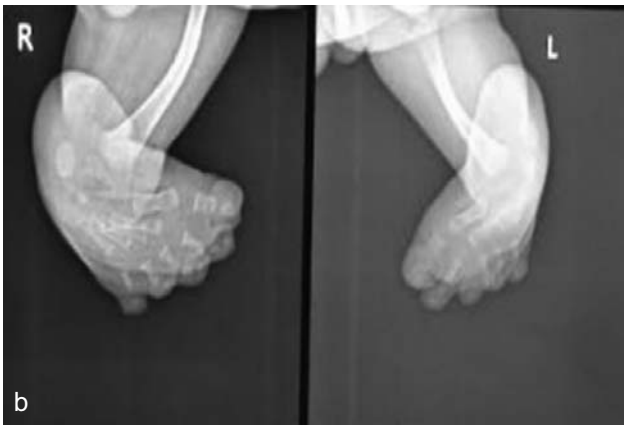


Figure 2a,b: Radiographs of the pelvis, lower limbs and feet demonstrated bilateral complete absent fibula, bowing of tibia and focal femoral deficiency of femur with pseudoarthrosis (Curved blue arrow) on each side **(Fig 2a).** Note the radiographic evidence of club feet marked with yellow arrows corresponds to clinically visible medially oriented forefeet **(Fig 2b)**.

Discussion

Fibular hemimelia is a congenital lower extremity anomaly. It is a tremendously rare disorder. There is a variation in its frequency. Only a handful of such cases are available in the literature with a reported incidence of approx. 5.7 to 20 cases per million births. Complete absence of fibula, aplasia /hypoplasia of fibula, and unilateral/bilateral involvement, all are encompassed in the same family. Unilateral fibular hemimelia is more common. The bilateral category occurs at a rate of 3:1.[1,3,4] This case showed a bilateral complete absence of fibula, a significantly rarer entity. Clinically, it presents at birth by limb length discrepancy or bowing of legs.[5] In this case, he presented with a similar presentation, bilateral short and bowed lower limbs were evident on clinical examination.

Its etiology remains unknown. Few theories explain that it is due to defects in the ectodermal ridge, one thought is that the amniotic band may affect limb bud growth.[6] Fibular hemimelia may occur sporadically or in association with proximal focal femoral deficiency, absence of lateral ray of toes or poly or syndactyly.[7] Literature also shows the association of maternal hyperpyrexia to congenital absent femur and fibular hemimelia.[8] Atcherman and Kalamchi classification is widely accepted which divides this condition into two types. Type 1 is characterized by minimal hypoplasia. Type II is characterized by a complete absence

of fibular hypoplasia.[9] In this case, there was a complete absence of fibula, so, according to Atcherman and Kalamchi's classification this case falls into type II fibular hemimelia. It was associated with bilateral proximal focal femoral deficiency with pseudoarthrosis, curved tibia and talipusequino-valgus deformity of feet or club feet. There was no evidence of absent lateral rays of feet , oligo, poly or syndactyly.

Fibular hemimelia may be detected antenatally by antenatal ultrasound, or anomaly scan. But our case presented post-natally and the mother couldn't get proper antenatal check-ups due to her low socio-economic status. It is usually further investigated with an X-ray pelvis and both lower limbs. Computed tomography may be suggested for a 3D view of limb lengthening for planning of surgical management. MRI may help in further characterization of pseudarthrosis of the femur.[10] This case was diagnosed based on a skeletal survey at the request of the primary physician, which showed a bilateral complete absence of a fibula associated with a bilateral absence of a focal part of the femur with false joints i.e. proximal focal femoral deficiency with pseudoarthrosis along with bilateral club feet on each side.

Treatment depends upon the specific symptoms. These cases usually undergo surgical procedures mainly limb lengthening procedures or amputation with a prosthesis.[11] This child was also referred to the orthopaedic team for further management as there were bilaterally shortened and bowed lower extremities owing to the already described type II fibular hemimelia diagnosed on the skeletal survey. Though, it is a non-lethal skeletal malformation but post-surgical residual deformity and disability persist which leads to aesthetic issues and is of social concern which demands patient as well as parental counselling as a part of management.

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Informed Consent: Informed consent was taken from parents of patient to publish the case including his clinical picture and X-ray images.

Conflict of Interest: None

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