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## Case Report

# Postaxial hypoplasia of the lower extremity (fibular hemimelia) presenting in a young adult male

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## ABSTRACT

Postaxial hypoplasia of the lower extremity, formerly termed as fibular hemimelia, is characterized by lower limb length discrepancy and a broad spectrum of anomalies involving the ipsilateral limb. It is a rare skeletal abnormality with an incidence of 5.7–20 cases per 1 million births. Herein, we present a young man with postaxial hypoplasia of the lower extremity who admitted to the hospital for a reason other than musculoskeletal complaints. While his limb length discrepancy was rather mild, the accompanying tarsal coalition was of an extensive form involving talus, calcaneus, navicular, and cuboid. Such extensive fusions of the hindfoot are very rare, and they are commonly associated with congenital syndromes such as postaxial hypoplasia of the lower extremity and Alpert syndrome. Therefore, further investigation for accompanying abnormalities is needed in cases with extensive fusions of the hindfoot.

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## Introduction

Postaxial hypoplasia of the lower extremity (PHLE), formerly termed as fibular hemimelia, is characterized by lower limb length discrepancy and a broad spectrum of anomalies involving the ipsilateral hip joint, femur, knee joint, tibia and fibula, ankle, and foot. Given the often associated abnormalities of the lower limb, the term postaxial deficiency may be more revealing than the term fibular hemimelia [1].

PHLE is rare, with an incidence of 5.7–20 cases per 1 million births [2]. The etiology of the disorder remains unclear, and the majority of the reported cases of PHLE develop sporadically without an evidence of an inheritable disorder or a family history of other birth defects. However, PHLE may present as a part of complex malformations such as, Femur-Fibula-Ulna syndrome, Thrombocytopenia-Absent Radius syndrome, Furhmann syndrome, Du Pan syndrome, or FATCO syndrome [3,4]. In two-thirds of patients with PHLE, unilateral involvement occurs. But in the cases which PHLE is a component of

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**Fig. 1 – Full-length standing anteroposterior radiograph of both lower extremities shows that the right lower limb is about 3 cm longer than its counterpart.**

a complex syndrome, the disorder is usually bilateral [2,4]. We herein present a case of a PHLE, who had a mild limb length discrepancy accompanied by unilateral extensive tarsal coalition and oligodactyly.

## Case report

A slightly limping 20-year-old man with no health complaints admitted to our hospital for mandatory health screening before military service. On detailed questioning, he declared a worsening left ankle pain. He had no family history of skeletal anomalies. Full-length standing anteroposterior (AP) radiograph of both lower extremities showed that his right lower limb was about 3 cm longer than its counterpart. The lengths of the right and left lower extremities were 106.9 and 103.5 cm, respectively (right femur: 55.7 mm, left femur: 55.1 mm, right tibia: 42.5 mm, left tibia: 41.4 mm). No skeletal abnormality was detected in the right lower limb (Fig. 1). Standing AP lumbar radiograph revealed a pelvic obliquity as the result of the limb length discrepancy. No spinal abnormality was noted on

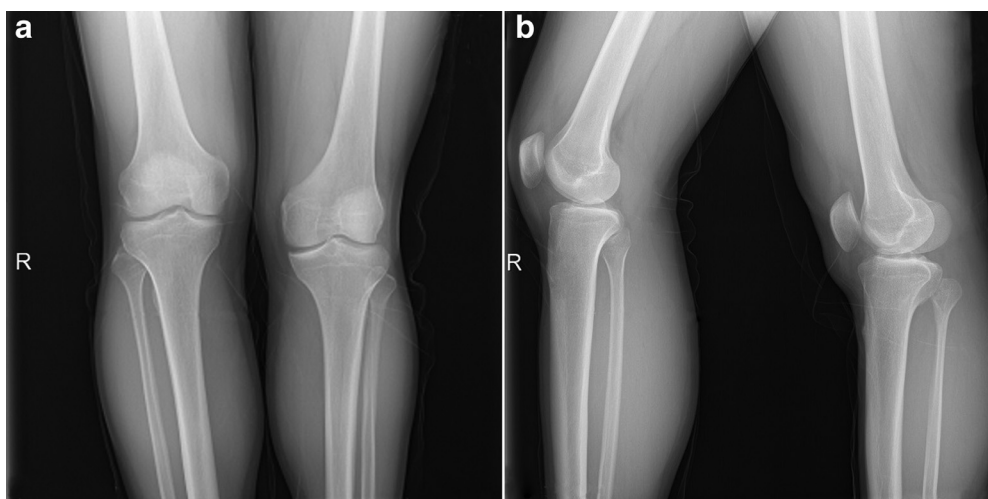


**Fig. 2 – Standing anteroposterior lumbar radiograph demonstrates a pelvic obliquity due to the limb length discrepancy. Note the sessile osteochondroma on the right superior pubic ramus.**

the lumbar radiograph (Fig. 2). On the standing AP and lateral (L) radiographs of both knees, no skeletal anomaly other than the evidence of length discrepancy between 2 lower limbs was observed (Fig. 3). AP left foot radiograph demonstrated oligodactyly with the deficiency of the fifth ray. Further, there were only 2 cuneiforms with the lateral cuneiform being absent (Fig. 4). L radiograph of the left foot depicted a tarsal coalition involving talus, calcaneus, navicular, and cuboid (Fig. 5). On AP and L radiographs of both ankles, the right ankle joint appeared normal. However, rounding of the talar dome and the corresponding concavity of the tibial plafond, consistent with the ball and socket ankle joint, were evident on the AP ankle radiograph. Distal portions of the right tibia and both fibulae were normal (Fig. 6).

## Discussion

The phenotype of PHLE has a wide spectrum of skeletal anomalies of the ipsilateral limb. Besides limb length discrepancy, the 10 features which PHLE may include are summarized in Figure 7 [5]. Among these features, the main problems which the patients with PHLE face are: limb length discrepancy, genu valgum, knee instability, tibial deformity, and foot and ankle deformities. The growth inhibition of the femur, tibia, and foot results in limb length discrepancy ranging from 2 to 25 cm. Genu valgus deformity which is observed in many patients with PHLE is also the result of the growth inhibition of the femur and tibia. A delay in the ossification of the lateral epiphysis of the proximal tibia may



**Fig. 3 – Standing anteroposterior and lateral radiographs of both knees show the length discrepancy between 2 lower limbs. Note the lateral translation of the left patella.**



**Fig. 4 – Anteroposterior left foot radiograph demonstrates oligodactyly with the deficiency of the fifth ray. Note there are only 2 cuneiforms with the lateral cuneiform being absent.**

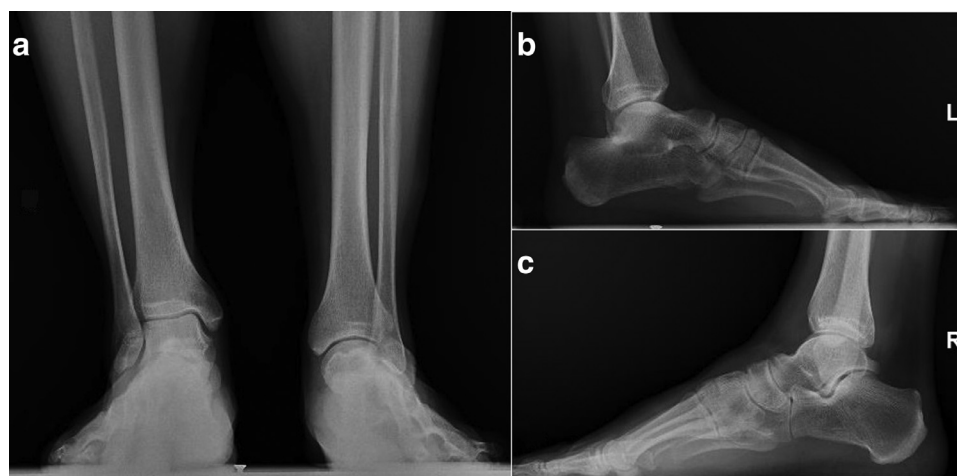


**Fig. 5 – Lateral left foot radiograph shows the tarsal coalition involving talus, calcaneus, navicular, and cuboid.**

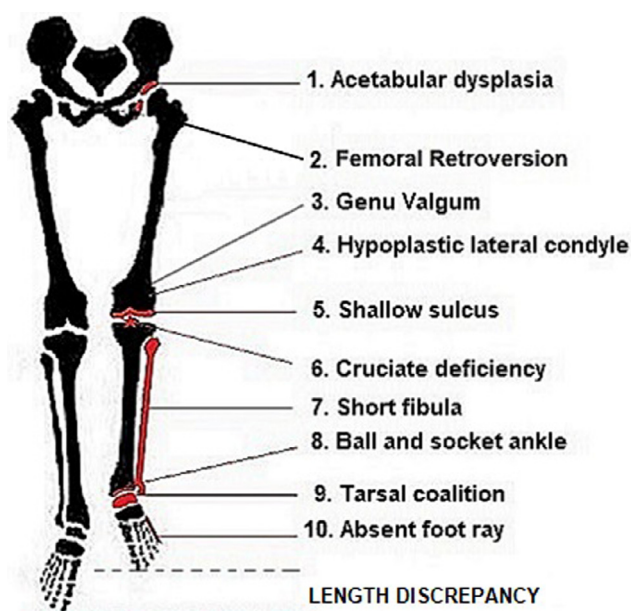
be observed in patients with genu valgus. The other problem concerning the knee joint is the anterior subluxation of the tibia which is the result of hypoplasia or aplasia of the anterior and/or posterior cruciate ligaments. Knee instability remains asymptomatic throughout young ages, however, as the child

grows and becomes heavier, the instability becomes increasingly symptomatic [3]. The patient we currently present was a young adult with a hardly distinguishable limp who has admitted to the hospital for a reason other than musculoskeletal complaints. He had a limb length discrepancy of only 3 cm, and had neither a knee joint disorder, nor a tibial deformity. Although his limb length discrepancy was of a rather mild form, the accompanying tarsal coalition was massive, causing ankle pain.

Foot and ankle deformities are reported to be the most disabling problems with PHLE. In many cases, there is distal tibial and talar dysplasia causing varying degrees of malformed and maloriented ankle joint surfaces. The concave distal tibial articular surface along with the rounded talar dome in the frontal plane are the parts of the anomaly termed as ball and socket ankle joint. Subtalar coalition is another deformity that may be included in the spectrum of anomalies involving the foot and ankle [3–5]. The coalition may be massive, and may



**Fig. 6 – Anteroposterior radiograph of both ankles (a), and lateral radiographs of the left (b), and the right (c) ankles. Rounding of the talar dome and a corresponding concavity of the tibial plafond, consistent with the ball and socket ankle joint, are evident on both the AP and L radiographs of the left ankle. Distal portions of the right tibia and both fibulae are normal.**



**Fig. 7 – Illustrative figure to demonstrate the list of the features which may be included in the phenotype of postaxial hypoplasia of the lower extremity.**

cause ankle pain as it does in our case. The radiographic imaging of our patient revealed the ball and socket ankle joint configuration along with the bony synostosis of talus, calcaneus, navicular, and cuboid. Such extensive fusions of the hindfoot are very rare, and they are commonly associated with congenital syndromes such as PHLE and Alpert syndrome [6]. The

deficiency of the 5th ray was also evident on the foot radiographs of our patient. The absence of 1 or more foot rays (the lateral ones) is a frequent finding in patients with PHLE [4].

The management of the patients with PHLE should consider both the functional and the cosmetic needs. Creation of a highly individualized reconstructive surgical plan is of great importance for management. The goal of this plan is to correct all of the deformities and deficiencies in the fewest number of surgeries spread throughout the growth years of the child. Surgical methods including limb lengthening, epiphysiodesis, amputation, or prosthetic rehabilitation are available in order to achieve equal limb length and functional hip, knee, and ankle joints [2,5].

Informed consent for publication: applicable.

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