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Case of incomplete fibular hemimelia with tarsal coalition, pes planus, ball and socket ankle

Inkomplet fibular hemimelia'ya eşlik eden tarsal koalisyon, pes planus, ball-socket ayak bileği deformitesi olgusu

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Abstract

Fibular hemimelia (FH) is a congenital disease with a clinical spectrum ranging from mild fibular hypoplasia to fibular aplasia. There is no proven genetic factor. Some anomalies can accompany FH such as tarsal coalition, ulnar hemimelia, amelia, syndactyly, several extremity anomalies, renal anomalies and cardiac anomalies. Our case is about unilateral and incomplete type of right-side FH in a 14 years old female patient. Tibia was curved (bowing) and short. Disparity of measure with left lower extremity was monitored. Tarsal coalition in osseous form, tibial curve anomaly and small bone part placed in fibula distal region compatible with FH, were visualized. There was curved joint form in the same ankle with hemimelia compatible with ball and socket ankle deformity. Calcaneal inclination angle was 120°. The findings were compatible with pes planus.

Keywords: Fibular hemimelia, Tarsal coalition, Fibular hypoplasia

Öz

Fibular hemimelia (FH), hafif fibular hipoplazi'den fibular aplaziye kadar uzanan klinik spektrumu olan bir konjenital hastalıktır. Kanıtlanmış bir genetik faktör yoktur. Tarsal koalisyon, ulnar hemimelia, amelia, sindaktili, kardiyak anomaliler, renal anomaliler ve çeşitli ekstremite anomalileri anomaliye eşlik edebilir. On dört yaşında kız olguda sağda tek taraflı ve parsiyel tipte FH mevcuttu. Tibia kavisli (eğimli), kısa ve ekstemiteler arasında uzunluk bakımından uyumsuzluk mevcuttu. Osseöz formda tarsal koalisyon deformiteye eşlik etmekteydi. Fibula distal parçası mevcuttu. Aynı taraf ayak bileğinde ball-socket deformitesi saptandı. Kalkaneal eğim açısı 120° idi. Bulgular pes planus ile uyumluydu.

Anahtar kelimeler: Fibular hemimelia, Tarsal koalisyon, Fibular hipoplazi

Introduction

Fibular hemimelia (FH) is a congenital disease characterized by the absence of a partial or complete part of the fibula. It occurs between 7/1000000 and 10/1000000 live births [1]. The word "Hemimelia" originates from the Greek word "Hemi and melos" meaning half limb because of shortening extremity [2]. This anomaly was defined by Gollier in 1698 for the first time. Stevens PM and Arms D [3] stated that this pathology is not only fibular deficiency but also accompanied by many deformities. Therefore, he emphasized that the terms FH or fibular aplasia, hypoplasia was insufficient to define this pathology and he defined this disease as postaxial hypoplasia of the lower extremity. This paper reports, with clinical and radiological findings, the case of a patient incomplete fibular hemimelia with tarsal coalition, pes planus, ball and socket ankle.

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Informed Consent: The author stated that the written consent was obtained from the parents of the patient presented in the study.
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Case presentation

Our patient is a 14 years old female who was followed for FH since childhood ages. The patient could walk but there was postural discrepancy and pain. Pain was mild range therefore no analgesic treatment was necessary. Abdominal ultrasound and cardiac evaluation were performed. Neither congenital anomaly nor variation was monitored. Upper extremities were normal. Disparity of extremities length was caused by curvature of the tibia (Figure 1). There was tarsal coalition in osseous form (Figure 2) and small part bone was visualized in the fibula distal region. There was no connection between tibia and hemimelic fibula part (Figure 3). In addition, calcaneal inclination angle was 120° which is compatible with pes planus. Furthermore, there was curved junction between tibia and talar bone corresponding to ball and socket ankle. The consent form was taken from the parents of the patient presented in this study.

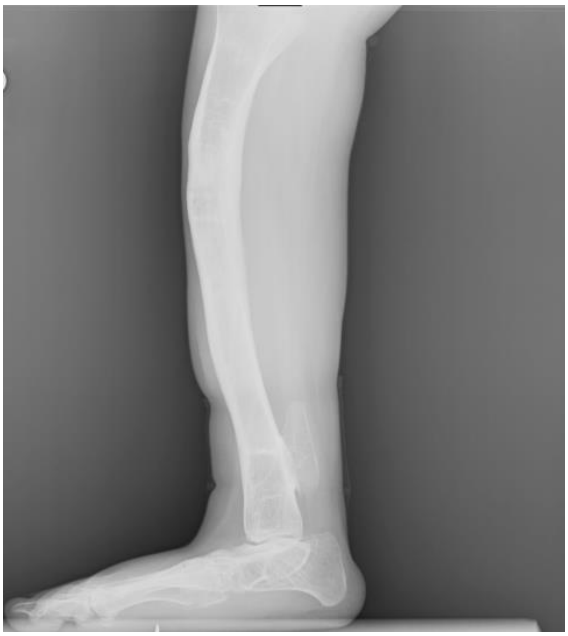

Figure 1: Curved tibia and tibial range was short

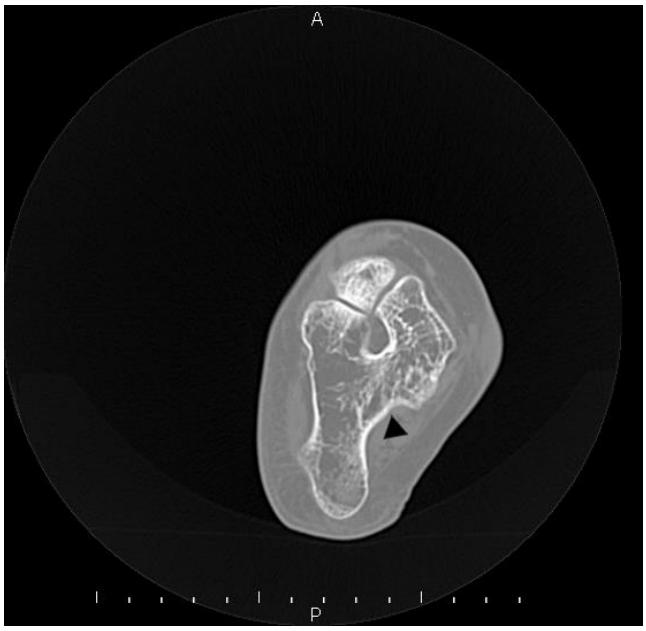

Figure 2: Arrow head: tarsal osseous coalition

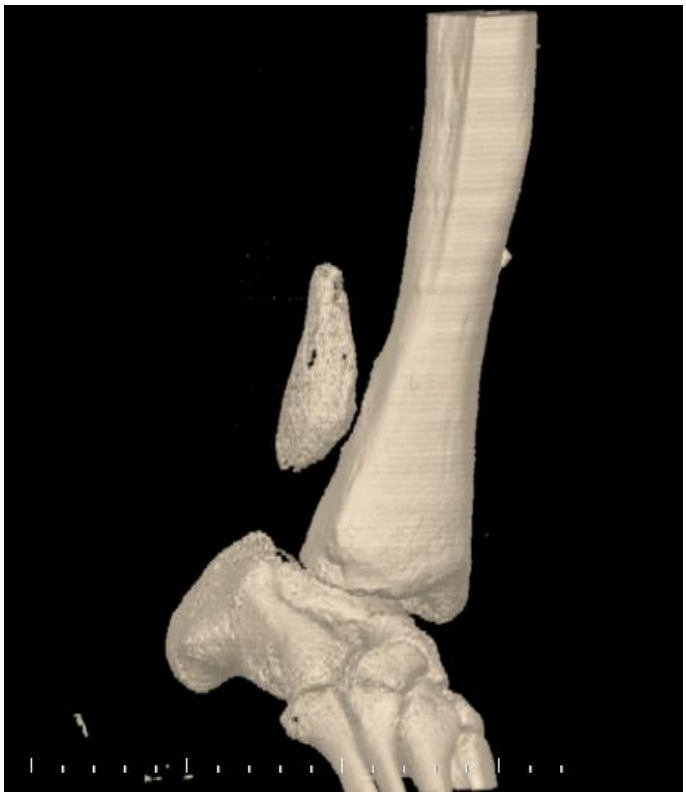

Figure 3: In 3D images hemimelic fibula and no connection with tibia

Discussion

FH 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 left [4]. The etiology of this anomaly, which is generally seen as unilateral, is not known exactly. No genetic predisposition was detected and most cases occurred sporadically [5]. It is seen twice more in boys than in girls [1].

There are various classification methods in literature. Most common used ones belong to Stanitski and Achterman-Kalamchy [1,2].

Three types of fibular hemimelia are described according to Stanitski [6]:

- Type I: It is mild type of FH. There is partial absence of fibula and tibial curving, without foot defect. Functional impairment is limited and bone defect is visible only on radiological examination.
- Type II: It is moderate type of FH. There is a complete or almost complete absence of the fibula. Lesion may be unilateral or sometimes bilateral. This type is linked with equinovalgus deformity of the foot and anteromedial bowing of the tibia. Bones of the foot may be hypoplastic or absent. There may be also tarsal coalition with development of a ball-and-socket ankle. There is moderate to marked limb shortening.
- Type III: It is severe type of FH. There is bilateral absence of the fibula. It may be associated with proximal femoral deficiency, deformities of the upper extremities or hemivertebra

Another classification according to Achterman and Kalamchi [7]:

- Type IA: Fibula is present. Epiphyses are small and mildest tibial growth defects are present
- Type IB: Partial absence of the fibula. Proximally 30% to 50% the fibula's length is absent. Distally, the fibula is present but does not support the ankle.
- Type II: Complete absence of the fibula

Our case can be classified as type II according to Stansky and between type IB and II according to Ackerman.

FH is a complex entity which has several anomalies including different organ and systems. The first visible sign of FH is the inequality in leg length. This may be due not only to the tibial shortness, but also to the proximal absence of the femur. The second important finding is bowing of the tibia in the anteromedial medial axis. This deformity in the sagittal and coronal plane may be accompanied by rotation. Most common anomaly associated to FH is tarsal coalition [1,8]. There was tibial bowing, leg length shortening and tarsal coalition with FH in our patient. Femur was normal.

Tarsal coalition is the fusion between the talus and calcaneus. The plain radiography is the most common diagnostic modality to visualize a tarsal coalition. However, tarsal coalitions can be completely cartilaginous. The radiographic diagnosis becomes more difficult. Computed tomography provides superior imaging capabilities, but it is not used routinely. A talocalcaneal coalition represents the failure of the embryonic precursors of the talus and the calcaneus to segment completely from one another. These coalitions are most often cartilaginous in young children. It can be visible only after progressive ossification [9].

The reported prevalence of tarsal coalition is ranging from 1% to 2%, however a recent cadaveric study demonstrated a prevalence of 13%. Middle facet talocalcaneal coalitions are noticed in 45% of all tarsal coalitions. Between the ages of eight and sixteen years, tarsal coalitions cause gradual flattening of the longitudinal arch and stiffness of the subtalar joint. Pain, which is present in about 25% of feet with a tarsal coalition, is the consensual indication for treatment [10]. Our patient presented a middle joint osseous type calcaneo-naviculer coalition. There was only mild degree of pain. No detected stiffness in subtalar joint but flattened longitudinal arc was present.

FH can accompany ulnar hemimelia, amelia, syndactyly, acetabular dysplasia, external rotational deformity, shortening varus/valgus femoral neck, hypoplastic lateral condyle, genu valgum, anterior cruciate ligament deficiency, posterior cruciate ligament deficiency, ball and socket deformity, valgus deformity, instability, dislocation, equinovalgus, tarsal coalition, absent rays, equinovarus, renal anomalies and cardiac anomalies [8]

Another important anomaly which was present in our patient is pes planus. It results from loss of the medial longitudinal arch and can be either rigid or flexible. Radiography is the main method for the evaluation. The longitudinal arch of the foot must be assessed on a weight bearing lateral foot radiography [11,12]. Inclination angle is low in pes planus (<20°) [13]. It is linked to ball-and-socket ankle deformity [14].

The ball-and-socket ankle joint is a malformation of the ankle in which the articular surface of the talus is hemispherical in both the anteroposterior and lateral projections. Congenital type of ball-and-socket ankle joint can be associate with tarsal coalition, short limb, ray fusion and deletion anomalies with or without FM [14].

FH is part of a multisystemic entity which also referred to postaxial hypoplasia. There are no proven genetic predisposing factors about this entity. There are sporadic cases with a few anomalies added to FH. That's why some aspect of FH is blurred. Our patient was admitted to hospital with mild symptoms and presented small part of the listed anomalies. Nevertheless, type of FH was not mild according to classification 'type IB'. In addition to discordance between clinical and radiological findings, our patient with FH presented not only a tarsal coalition which is a common anomaly but also rare deformities like pes planus and ball and socket ankle.

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