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Congenital Fibula Aplasia, Tibial Campomelia with Oligosyndactyly in an Infant (Fatco Syndrome): Ragiographic Findings and A Case Report

[1] Shamaki AMB, [2] Sule MB, [3] Erinle SA, [4] Gele IH, [5] Shafiu H, [6] Abdullahi A

[1, 2] Department of Radiology, Usmanu Danfodiyo University, Sokoto, Nigeria
[3] Department of Radiology, Federal Medical Centre Bida, Niger State, Nigeria
[4, 5] Department of Radiology, Usmanu Danfodiyo University Teaching Hospital, Sokoto, Nigeria
[6] Department of Paediatric, Usmanu Danfodiyo University Teaching Hospital, Sokoto, Nigeria

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Corresponding Author: **Sule MB**

Abstract

FATCO syndrome is a rare congenital limb malformation characterized by absence or hypoplasia of the fibula, aplasia or dysplasia of the tarsal bones, shortened or curved tibia (campomelia) and oligosyndactyly (missing or fused digit especially in the feet. The etiology is unknown but sporadic mutations are most likely with no consistent genetic pattern confirmed.

This is a three month old female infant that was referred for plain radiograph of the right leg on account of limb-length shortening and lateral deviation of the foot. The plain radiograph demonstrated complete absence of the right fibula (Fibula aplasia), shortening with anterior bowing of the tibia (campomelia), and absence of the 3rd and 4th metartasals and digits with fusion of the demonstrated 2nd and 3rd digits (oligosyndactyly). There is associated valgus deformity of the right ankle joint and foot. The contralateral left leg had normal appearances.

The presence of the above findings prompted the diagnosis of **FATCO** syndrome in a three month old female infant, hence the presentation of this case due to its rare occurrence, peculiar presentation and to review the literature.

Keywords: Campomelia, Fibula Aplasia, Tarsal Aplasia, Plain Radiograph

Introduction

FATCO syndrome is a rare occurrence characterized by fibula aplasia, tibial campomelia and oligosyndactyly, this was first described in 1981 by Hecht and Scott.[1, 2]. This syndrome is suspected to occur in a sporadic pattern, though seen in cases with autosomal dominant or X-linked inheritance [1, 3].

Congenital deficiencies involving the limbs are relatively common birth defects with a prevalence of about 1 in 2000 neonates and characterized by aplasia or hypoplasia of the bones of the limbs. The disease spectrum ranges from mild fibula hypoplasia to fibula hypoplasia, the combination of fibula aplasia, tibial campomelia and oligosyndactyly constitute the FATCO syndrome which is purely descriptive terms for a syndrome of unknown genetic basis and inheritance [4, 5].

Affected cases of FATCO syndrome do have a normal mental development without any facial dysmorphism or other associated anomalies, this has being found to be of utmost importance in counselling of the patients relations [4, 6-8]. The combination of fibula hypoplasia, tibial campomelia and oligosyndactyly was termed variant of FATCO syndrome, this was described by Goyal and colleagues [4, 9].

FATCO syndrome is most common in males and diagnosed postnatally, and most often the lower limbs are commonly affected and a few cases may have involvement of the upper limbs cocurrently [10, 11].

Surgical management is the mainstay of treatment in patients with FATCO syndrome; this is meant to achieve foot preservation and equalization of the length of extremeties, and include limb-lengthening procedures, epiphysiodesis, Syme's or Boyd's amputation with prosthesis [4, 12].

Case Report

This is a three month old female infant that was referred for plain radiograph of the right leg on account of limb-length

shortening and lateral deviation of the foot.

The patient is the third child of a 35-year-old woman who claimed to have had adequate antenatal visit with appropriate medication, no history of similar occurrence in the siblings or family.

The patient is conscious and alert with good neck control, good suckling reflex, with some limb-length discrepancy, lateral deviation of the ankle and foot with fused distal two digits.

The plain radiograph demonstrated complete absence of the right fibula (aplasia), anterior bowing of the tibia (campomelia), and absence of the 3rd and 4th metartasals and digits with fusion of the demonstrated 2nd and 3rd digits (oligosyndactyly). There is associated valgus deformity of the right ankle joint, abnormal orientation of the talus and calcaneum when compared to the left foot (Figure 1&2). The contralateral left leg, ankle and foot had normal appearances (figure 3).

The presence of the above findings prompted the diagnosis of **FATCO** syndrome in a three month old female infant; the parents were advised to consult a paediatric surgeon for expertise management. FATCO syndrome happens to have a rare occurrence and peculiar presentation hence the need for this report and to review the literature.

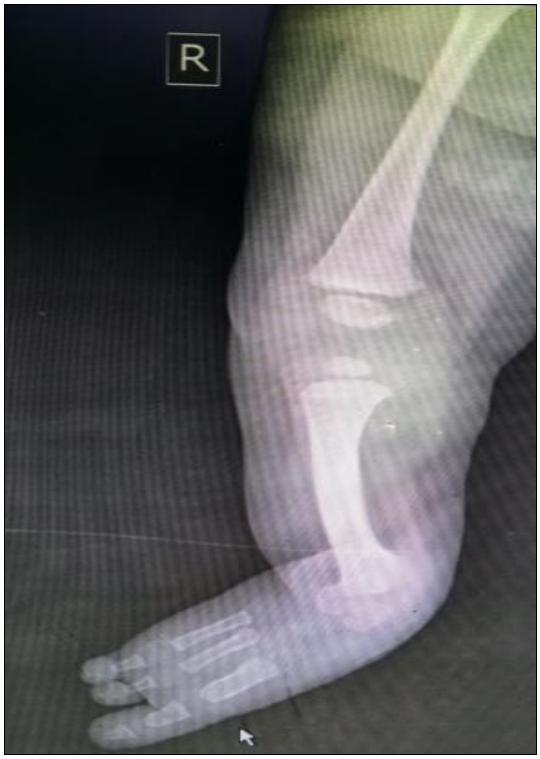

Fig 1: Plain radiograph of the right limb demonstrating a normal femur, shortened tibia with some anterior bowing (campomelia), complete absence of the fibula (aplasia), absent 4th and 5th digit with fused 2nd and 3rd digit (oligosyndactyly); these comprise the **FATCO** syndrome. Vagus deviation of the right ankle and disorientation of the talus and calcaneum relation are also demonstrated

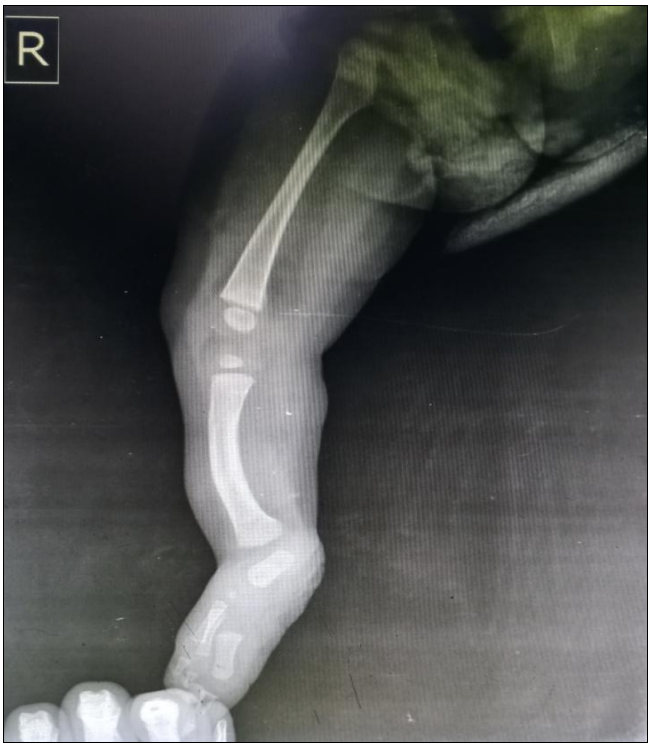

Fig 2: The tibial bowing (campomelia) is better demonstrated; the increase in the skin-fold thickness is also demonstrated. The other components of the FATCO syndrome; fibula aplasia and oligosyndactyly are also demonstrated

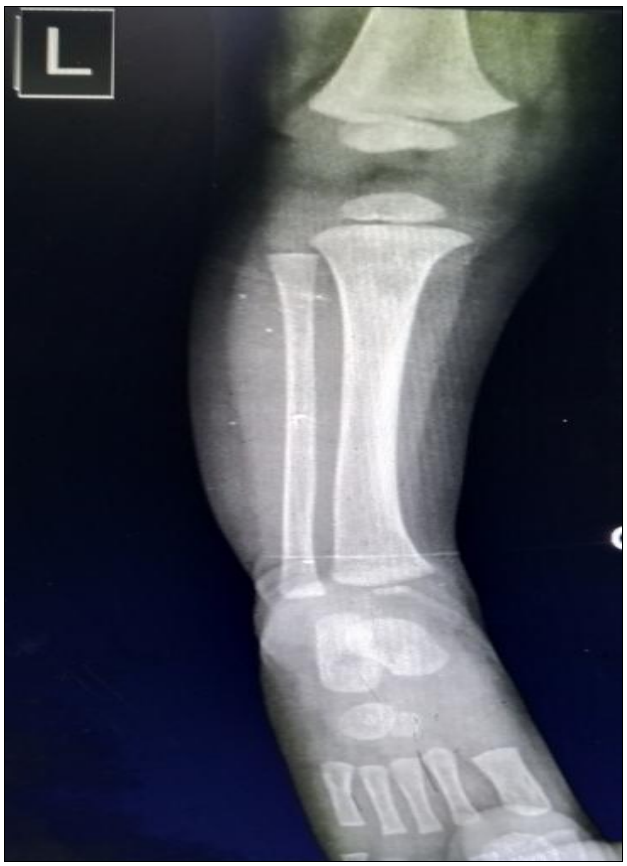

Fig 3: Plain radiograph of the left leg demonstrating part of the femur, normal tibia and fibula, complete tarsal bones and complete digits for the patient's age

Discussion

FATCO syndrome is a rare occurrence characterized by fibula aplasia, tibial campomelia and oligosyndactyly, this was first described in 1981 by Hecht and Scott. [1, 2]. The case under review had fibula aplasia, tibial campomelia and oligosyndactyly; hence termed having FATCO syndrome and agreeing with these literatures.

FATCO syndrome is suspected to occur in a sporadic pattern, though seen in cases with autosomal dominant or X-linked inheritance [1, 3]. The case under review has no family history of similar occurrence and a female; this is most likely sporadic in appearance and not likely an X-linked inheritance.

Affected cases of FATCO syndrome do have a normal mental development without any facial dysmorphism or other associated anomalies, this has being found to be of utmost importance in counselling of the patients relations [4, 6-8]. The case under review had normal mental and milestone development for her age, had no facial dysmorphism and associated anomalies, thereby agreeing with these literatures.

The combination of fibula hypoplasia, tibial campomelia and oligosyndactyly was termed variant of FATCO syndrome, this was described by Goyal and colleagues [4, 9]. The patient under review had fibula aplasia and other components of the syndrome and not a variant of the syndrome, thereby conforming to these literatures.

FATCO syndrome is most common in males and diagnosed postnatally, and most often the lower limbs are commonly affected and a few cases may have involvement of the upper limbs cocurrently [10, 11]. The patient for this review happens to be a female infant with postnatal diagnosis and had no affectation of the upper limbs.

Radiographic examination with other imaging techniques such as prenatal ultrasonography, computed tomography and magnetic imaging resonance imaging are also important in demonstrating the components of FATCO syndrome [4, 11, 13]. The case under review was diagnosed following plain radiographic examination in conforming to these literatures.

Surgical management is the mainstay of treatment in patients with FATCO syndrome; this is meant to achieve foot preservation and equalization of the length of extremeties, and include limb-lengthening procedures, epiphysiodesis, Syme's or Boyd's amputation with prosthesis [4, 12]. The parents of the case under review were advised to consult the services of a paediatric surgeon for expertise management, and most likely may fall within the aforementioned mainstay of treatment.

Conclusion

Limb anomalies are relatively rare, when suspected plain radiographs can demonstrate the affected portions of the limbs and hence establishing a diagnosis for prompt onset of management in affected cases.

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