

A case report on fibular aplasia, tibial campomelia, oligosyndactyly syndrome variant in a male infant

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Abstract

Fibular aplasia, tibial campomelia, and oligosyndactyly (FATCO syndrome) is a rare, genetic, congenital limb malformation characterised by unilateral or bilateral fibular aplasia, tibial campomelia, and lower limb oligosyndactyly involving the lateral rays.

A newborn male born at term via a Caesarean Section presented with malformations consisting of tibial campomelia, unilateral fibular hypoplasia, and oligosyndactyly, a "FATCO variant" case. On radiographic examination, an anterolateral shortened and bowed right lower limb at the distal third of the tibia, a rudimentary right fibula and absence of three rays on right foot were revealed.

"FATCO syndrome" although rare may be linked to involvement of different body systems with morbidity and mortality. Proper parent counseling is a key aspect of this syndrome. Timely diagnosis and management with a multidisciplinary approach is essential to avoid lifelong disability, which can be a hurdle in a developing country.

Keywords: Fibular Hypoplasia, Oligosyndactyly, Tibial Campomelia, FATCO.

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Introduction

Congenital limb deficiency which is characterized by hypoplasia of bones is a common birth defect occurring in 1/2000 neonates.¹ The absence of fibula with an estimated incidence of 5.7 to 20 cases per 1 million births suggests that this is a very rare occurrence.² Despite that, it still is the most common deformity amongst the long bone deficiency conditions.³ 'Fibular Hemimelia' was first explained by Gollier in 1698.⁴ The exact cause remains uncertain, though it is probably thought to be sporadic since most of the cases are secondary to teratogens and radiations.⁵ The term fibular hemimelia covers a variety of disorders ranging from moderate fibular hypoplasia to

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fibular aplasia.⁴ It can be part of a malformation syndrome or as an isolated condition.³ It is characterized by valgus deformity, knee, and ankle anteroposterior instability, clubfoot, the tarsal coalition with the deficiency of lateral rays of the foot, tibia and femur shortening, and flexion contracture.⁶ In a literature review by Courtens et al, five cases on fibular aplasia having similar features were studied and three findings oligosyndactyly (without heart defect), tibial campomelia and fibular agenesis were common to all hence the name Fibular Aplasia — Tibial Campomelia — Oligosyndactyly (FATCO) syndrome.⁵ The main principal behind managing such patients is limb preservation and elongation in order to equalize both limbs which can be done via surgical correction and prosthesis use. If not done promptly, it could lead to lifelong disability.⁷ We report the case of a male newborn presenting with fibular hypoplasia, tibial campomelia, and oligodactyly on the first day after birth from a Low-middle income country where diagnosis and management of such cases can be a challenge. Different congenital limb deficiencies have been observed in some studies in Pakistan but according to the best of our knowledge, this is the first case of "FATCO syndrome" which is reported from Pakistan.^{8,9}

Case Report

A male neonate born via elective Caesarean section at 38 weeks was identified with fibular hypoplasia, tibial campomelia, and oligodactyly on the first day of birth at the Aga Khan Secondary hospital in February 2020. All reported antenatal scans were unremarkable. He was the 5th pregnancy of a healthy mother, who had no history of prenatal complications, antenatal drug usage, or any other comorbidities like diabetes or hypertension. The infant's birth weight was 2580 g (50th centile), the birth length was 50 cm (50th to 70th centile) and the head circumference at birth was 34 cm (50th centile). There was no family history of limb abnormalities and the only significant aspect was a consanguineous marriage. The baby was kept under observation in the nursery for 6 hours. The family belonged to a middle socio-economic household from an urban area.

Physical examination revealed shortened and bowed right lower limb at the distal third of the tibia (Figure-1a).

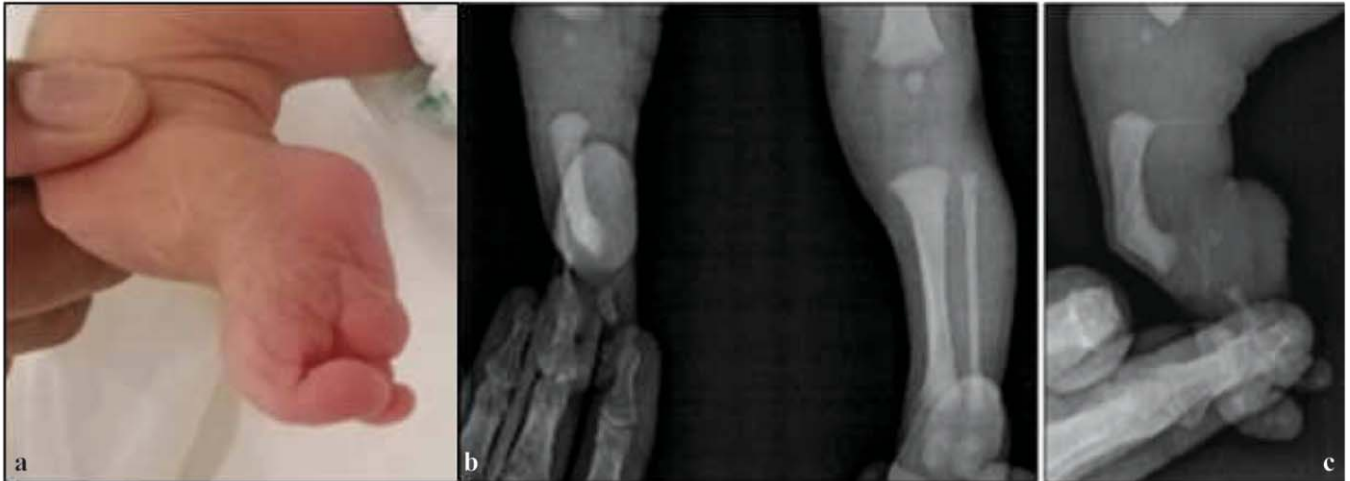


Figure-1: a) Shortened and bowed right lower limb at the distal third of the tibia. b, c) Radiological image showing short and bowed right tibia with pointing of the distal end along with rudimentary right fibula.

Only two toes were found on the right extremity with absent middle 3 toes whereas the left lower limb was normal. There was no associated facial dysmorphism or upper limb abnormality. No findings suggesting congenital hip dysplasia were found on hip ultrasound.

On radiological examination, right tibia was short and bowed with pointing of the distal end (Figure-1b, 1c) and right fibula was rudimentary. Only 2 digits were visible on the right foot while other three rays were absent. Both femurs and the left fibula and tibia were normal with all 5 rays evident in the left foot. This fibular agenesis was type II according to Achterman and Kalamchi classification¹⁰ and type IIIH4 according to Stanitski classification.¹¹

The parents were informed about the nature of the deformity and given follow up with the Orthopaedic consultant for the treatment options. A possible surgical management with the fabrication of a new orthosis was also proposed and included a follow up in 3-6 months to review limb length. Follow up with a Geneticist was also advised for future pregnancies. Unfortunately, the patient was lost to follow up.

Discussion

To the best of our knowledge, this is the first case of "FATCO syndrome" that is reported from Pakistan. Around 20 cases discussing "FATCO syndrome" have been previously reported in the literature with high clinical heterogeneity amongst them. All cases had lower limb involvement while upper limb involvement was reported in around two-third of cases.

As seen in all previously mentioned patients, there was a male sex predilection with a male to female ratio of 4:1.¹² Our case correlates to the radiological classification of a

variant of "FATCO syndrome" as stated by Goyal et al, who, in addition to all previously reported 13 patients with fibular aplasia, added a case of unilateral fibular hypoplasia, oligodactyly, and tibial campomelia. He termed this as a variant of "FATCO syndrome" with all 3 findings.³

Similar to our case, no facial dysmorphism, decreased mental growth or other abnormalities have been observed in children with "FATCO syndrome", which is one of the most significant aspects in counseling the patient's family. The only exception was a case reported by Kitaoka et al. in which the Japanese male patient had an additional cleft lip.¹³

The genetic basis of this disease is still unclear. A few genes including WNT7a, TP63, and WNT10B were analyzed in previous cases for a possible mutation causing "FATCO syndrome" but all were found to be normal.¹² Supporting these findings, a case by Yilmaz et al. reports a female monozygotic twin with "FATCO syndrome" having a normal female twin. Since monozygotic twins have the same DNA, it could be said that "FATCO" is not only due to DNA mutations but other mechanisms also play an important role.¹⁴ Since our patient had no significant perinatal history, any family history of limb abnormalities, or any similar condition in any of the four siblings, this suggests that the syndrome occurred sporadically. Similarly, the mode of inheritance of this disease is also unclear. The syndrome is sporadic and is either X-linked or autosomal dominant with decreased penetration, as stated by Biegansky et al.¹² Still, more information is required to completely understand the condition.

Treatment is surgical and involves amputation,

epiphysiodesis, and limb-lengthening procedures with a prosthesis which is typically individualized from one case to another. The most essential goal of treatment is to preserve the foot and equalize the length of the extremities. In cases where there is a non-functional foot and more than 50 percent shortening at birth relative to the estimated length, Syme's or Boyd's surgical amputation with early use of a prosthesis is recommended. Best outcomes are achieved and accepted by the patients when a combination of epiphysiodesis and elongation is done.⁷

Conclusion

We report the first case in Pakistan as per our knowledge. Still there is a lot that we don't know regarding "FATCO syndrome". Parental counseling and educating them for what is inevitable and for future pregnancies along with psychological support for parents and children is one of the key elements in management as was planned in our case.

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Conflicts of Interest: There is no conflict of interest.

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Availability of Data and Material: Case details are not publicly available because the data is patient medical records but are available from the corresponding author on reasonable request.

Parent Consent: Informed consent was taken from the parents prior to writing the manuscript, for publishing the case.

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