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FATCO Syndrome (Fibular Aplasia, Tibial Campomelia, Oligosyndactyly with Talar Aplasia). A Case Study

Khurshid Ahmad[(E)], Hilal Ahmad Malla[(F)], Sheikh Dawood[(B)]

Deptt. of Orthopaedics, Govt Medical College Jammu, J&K, India

SUMMARY

FATCO syndrome consists of fibular hemimelia, tibial campomelia and oligosyndactyly. FATCO syndrome can also be associated with other congenital anomalies; therefore, every case needs thorough evaluation so as to make the management of the patient easier. A few cases of this syndrome have been described in literature but only two cases have been reported in India so far. We present a 3-year-old male child born of a non-consanguinous marriage with FATCO syndrome and ipilateral talar aplasia without any other congenital anomalies.

Key words: fibular hemimelia, absent talus, tibial campomelia, oligosyndactyly

BACKGROUND

FATCO syndrome consists of fibular hemimelia, tibial campomelia and oligosyndactyly. FATCO syndrome can also be associated with other congenital anomalies; therefore, every case needs thorough evaluation so as to make the management of the patient easier. A few cases of this syndrome have been described in literature but only two cases have been reported in India so far. We present a 3-year-old male child born of a non-consanguinous marriage with FATCO syndrome and ipilateral talar aplasia without any other congenital anomalies.

CASE REPORT

A 3-year-old male boy born to a 26-year-old housewife and a 28-year-old father presented to our orthopaedic OPD with complaints of a short right lower limb with ipsilateral short and deformed ankle and foot. The boy was the first child of a non-consanguinous marriage, born at full term by a caesarean section. The pregnancy and delivery periods were uneventful. The pre- and post-natal periods were without any complications. There was no delay in developmental milestones. There was no history suggestive of hypertension, diabetes mellitus, any other chronic illnesses or any congenital anomaly in the family.

On examination the child had a short right lower limb with a skin dimpling over the anterior aspect of the mid-third of the right leg. There was a valgus deformity at the ankle with a short right foot and absent lateral rays (Fig. 1). Segmental measurements revealed a shortness of 4 cm in right leg. The child had no joint contractures or other abnormalities in the upper and lower extremities.

Radiographs of the concerned limb showed anterior bowing of the tibia and aplasia of the fibula and talus. Three lateral rays were also absent in the right foot (Fig. 2 and 3).

The parents of the child were informed regarding this syndrome and the possible treatment options, including the limb reconstruction.

DISCUSSION

FATCO syndrome, consisting of fibular aplasia, tibial campomelia and oligosyndactyly is a rare entity. Patients with this syndrome present with a short leg, a deformed and short ankle and foot, absent rays and a bent tibia [1,2]. A few cases of this syndrome have already been described in the literature. In India, only two cases have been reported so far [3]. Fibular aplasia is the most common congenital aplasia of long bones, followed by tibia, ulna, radius and femur [4,5]. Its incidence is 5.7 to 20 per 1 million births [6]. It may be an isolated abnormality or may be associated with other anomalies [7]. Evans et al [8] has differentiated fibular aplasia with ectrodactyly from FATCO syndrome as the latter is a heterogeneous disorder with a dominant inheritance. Kumar et al. [9] has described Fuhrmann syndrome, which includes aplasia/hypoplasia of the ulna, pelvis, femur, fibula and digital abnormalities with nail dysplasia. Our case had no such abnomalies.

The treatment of FATCO syndrome includes the use of orthoses, limb lengthening, epiphysiodesis, early amputations and application of a prosthesis. Most series have reported good results with Syme amputation [10]. In our case, the concern to the parents was the short limb and weight of the orthosis.

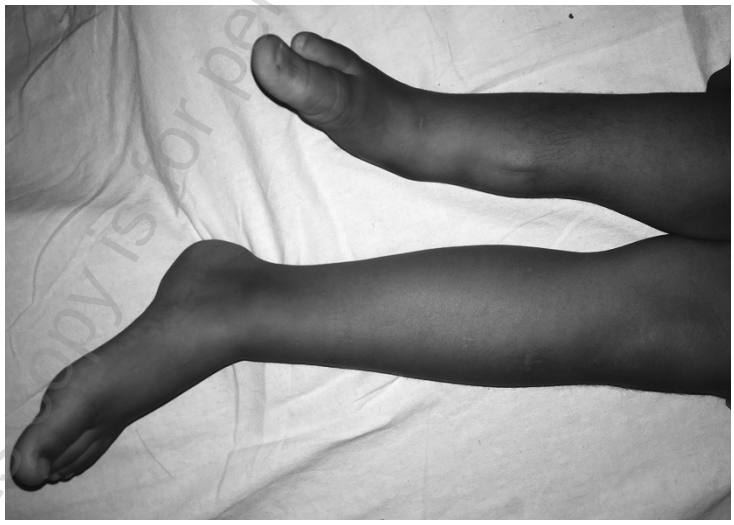

Fig. 1. Clinical photograph showing short leg with a dimple over anterior aspect of right leg, valgus at ankle and absent lateral rays

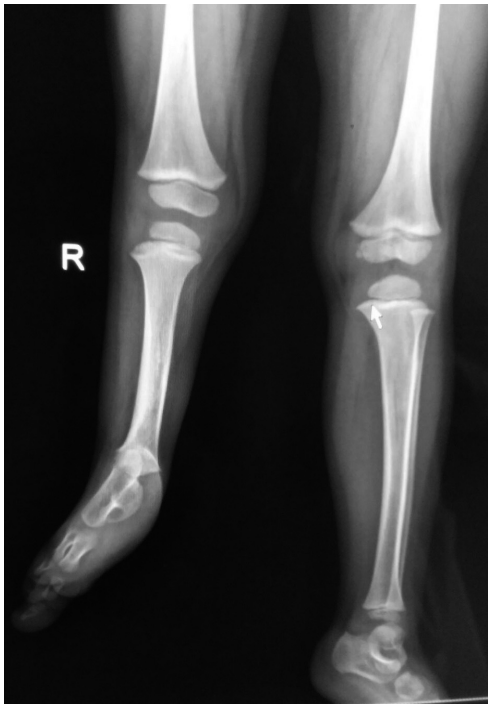

Fig. 2. Anteroposterior radiograph of right leg and ankle showing absent fibula, talus and lateral rays on right side

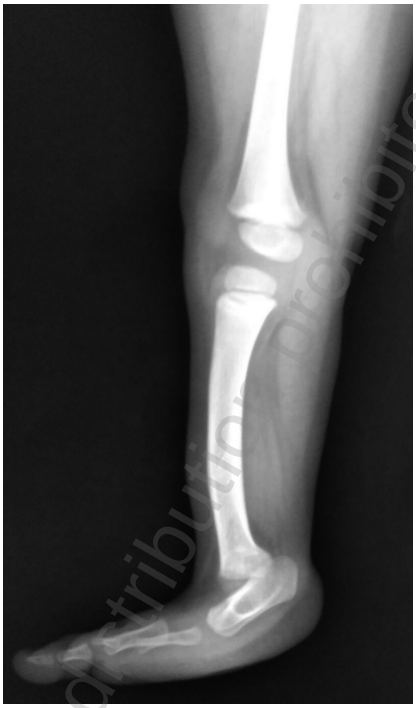

Fig 3. Lateral radiograph of right leg and ankle showing anterior bowing of tibia and absent talus

They did not agree with any treatment options other than an orthosis.

Although FATCO syndrome can occur both in males or females, the previously reported cases had a male predominance like our case. As per our literature review, although a few cases have already been reported. in India only 2 cases have been reported so far [3]. Fibular hemimelia patients should be thoroughly evaluated as it can be associated with other malformations like tibial pseudoarthosis, proximal femoral deficiency, pelvic hypoplasia/aplasia, digital and facial anomalies. Parents need proper counselling regarding the nature of the disease, its treatment methods and prognosis.

CONCLUSION

FATCO syndrome, though a rare entity, should be kept in mind while evaluating cases of fibular hypoplasia. This syndrome is associated with considerable psychosocial morbidity and mortality. Therefore a timely genetic counselling, early diagnosis and treatment can prevent or reduce the burden of complications of this syndrome.

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Adres do korespondencji / Address for correspondence
Khurshid Ahmad D-11 Medical Enclave Govt Medical College Jammu.
Pin 180001. Phone: 0191-2549311, Mobile no. (+919596583527
e-mail: bhatkhurshid79@gmail.com

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