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Case Report

# A Rare Syndrome in the Newborn: Fatco Syndrome

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## Abstract

In our case, a newborn with FATCO (fibular Aplasia, Tibial Campomelia and Oligodactyly) syndrome is described. FATCO syndrome is a syndrome characterized by shortening and forward bending of the lower extremities at the distal third point of the tibia, standing oligodactyly and fibular aplasia. In our case, there was no other anomaly except extremity anomalies. The karyotype was normal male (46,XY). In addition to presenting a new case report of this syndrome, we would like to emphasize the importance of prenatal diagnosis and genetic counseling for families at high risk for genetic disease, especially in developing countries.

**Keywords:** fatco syndrome; fibular aplasia; tibial campomelia; oligodactyly

## Introduction

Fibular hemimelia is the rarest congenital anomaly of the extremity, with malformation of the tibia and/or toes, or complete absence of the fibula, from mild fibular hypoplasia to complete fibular aplasia. FATCO syndrome (MIM#246570), which was first described by Hecht and Scott in 1981, is an extremely rare syndrome that regulates fibular aplasia, tibial campomelia, and oligosyndactyly by physical examination organs. The primary common effect of the syndrome is fibular aplasia, tibial campomelia, and oligosyndactyly [1]. Although the syndrome is thought to be sporadic, it is present in cases with autosomal dominant or X-linked inheritance [2]. Work up to accumulate nearly a hundred cases in the literature. It is seen that the neurodevelopment of the past cases is normal. In the published cases, micrognathia, hypertelorism and hydrocephalus are among the protective craniofacial anomalies [3]. We would like to discuss the case who underwent FATCO syndrome examination in the physical practice in the light of the literature.

## Case Report

The patient, who was born at 38 weeks by elective cesarean section from a 28-year-old mother, was hospitalized in the neonatal intensive care unit for examination and treatment due to extremity anomaly. The prenatal follow-up of the patient was unremarkable. In the first physical examination findings, body weight was 3230 g (25-50 p), head circumference was 35.2 cm (25-50 p), and height was 46 cm (3-10 p). Physical examination revealed fibular aplasia in the right lower extremity, shortening of the tibia and anterolateral bowing, as well as soft tissue dimpling and right foot oligosyndactyly. Four toes (without the fifth metatarsal) were found on the right foot (Figure 1a/1b). There was tibial campomelia and fibular aplasia on the direct X-ray of the patient (Figure 2). There were no associated abnormalities in the upper extremities. Abdominal and cranial ultrasonography and echocardiography of the patient were normal. The patient's chromosome analysis was 46,XY. WNT7A gene analysis was performed in the differential diagnosis and it was normal. The patient was diagnosed with FATCO syndrome with the present physical examination findings.

**Figure 1a.** Image of short and campomelic tibia

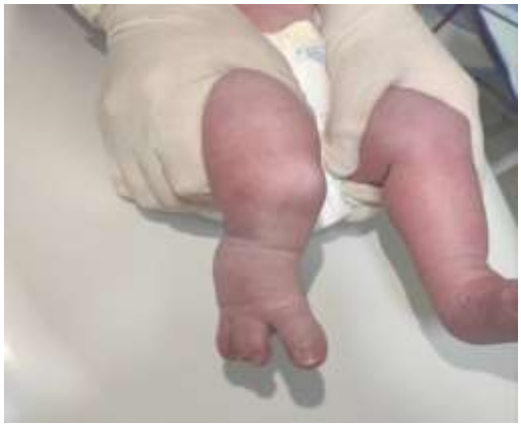

**Figure 1b.** Soft tissue dimpling and right foot oligosyndactyly

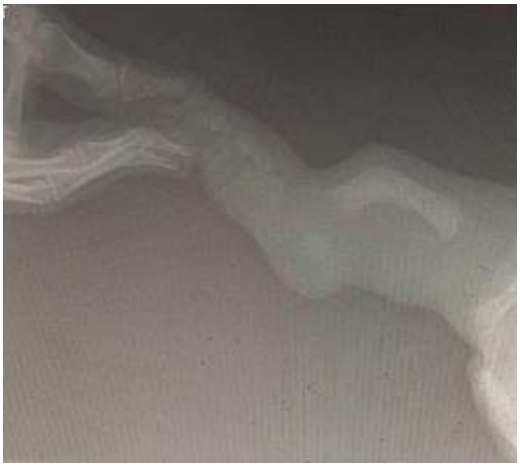

**Figure 2.** Direct radiograph of short campomelic tibia and fibular agenesis

## Discussion

Congenital limb deficiencies are common birth defects that occur in 1 in 2000 newborns and are characterized by aplasia or hypoplasia of the limb bones [4]. Fibular hemimelia (FH), first described by Gollier in 1968, is a rare congenital anomaly. FH is mostly seen unilaterally, isolated and sporadically for an unknown reason [5]. However, FH may be part of a malformation syndrome. Possible components include shortening of the femur and tibia, clubfoot, valgus deformity, flexion contracture, and anteroposterior instability of the knee and ankle, as well as tarsal coalition with lack of lateral rays of the foot. Although fibular hemimelia is rare, the most common long bone deficiency is malformation [6].

Approximately one hundred cases with the diagnosis of FATCO syndrome have been reported in the literature. The definition of the syndrome was first proposed by Courtens et al in 2005. The diagnosis was mainly based on the locations and types of malformations [5]. The main common findings in the cases described by Hecht and Scott in 1981 and the case of Courtens et al., and in all five patients, were fibular aplasia, tibial campomelia, and oligosyndactyly. That's why they suggested calling it the fibular aplasia-tibial campomelia-oligosyndactyly (FATCO) syndrome. In our case, all malformations were present.

Fuhrmann syndrome, Al-Awadi syndrome, Du Pann syndrome and FFU (Femur-Fibula-Ulna) syndrome should be excluded in the differential diagnosis of FATCO syndrome. FFU (Femur-Fibula-Ulna) syndrome was excluded because the patient did not have short femur and the ulna was

normal. Du Pann syndrome (also known as acrosomesomelic dysplasia) is characterized by the presence of fibular aplasia and brachydactyly. In Fuhrmann syndrome, femoral curvature is seen, not tibial. In addition, Fuhrmann syndrome was excluded by analysis of the WNT7A gene [2,5]. Renal anomalies such as extremity/pelvic hypoplasia/aplasia, horseshoe and polycystic kidney, and abnormal facial features such as cleft palate, hypertelorism and micro-retrognathia seen in Al-Awadi syndrome were excluded in our case [7].

The mental development of children with FATCO syndrome is normal and they do not have facial deformities or other anomalies, which is important for counseling the patient's family [8,9]. So far, two patients have been diagnosed prenatally. In one case, nuchal translucency was found as the earliest finding, which may guide ultrasonographic examination for early prenatal diagnosis [10,11].

In conclusion, fibular aplasia and tibial campomelia, the major components of FATCO syndrome, have been reported in all patients diagnosed to date. In most cases, oligosyndactyly is seen in the hands or mostly in the feet. While counseling parents, it should be explained that these children will show a normal neurodevelopment.

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