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IMAGES IN CLINICAL PRACTICE

A NEWBORN WITH LOWER LIMB DEFORMITY IN TIBIA AND FIBULA, AND OLIGODACTYLY

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A female neonate born at 34 weeks of gestation by cesarean section from a 28-year-old healthy mother with gravity 5 and parity 5 was admitted to the neonatal intensive care unit (NICU) for further evaluation and treatment due to an anomaly in her lower extremity. Apgar scores were 4 and 6 at the 1st and 5th minutes. Antenatal history revealed irregular pregnancy follow-up without fetal anomaly on obstetric ultrasound. There was no maternal history of teratogen exposure, except for 1-2 cigarettes per day throughout the pregnancy. There was no kinship between the parents, the other children were alive and healthy. Parental history revealed methamphetamine and marijuana use for the last ten years. On examination, the baby weighed 2350 gm (25-50th centile), height was 46 cm (50th centile) and head circumference was 32 cm (25-50th centile). Physical examination was normal except for the bowed and short left leg, and oligodactyly (3 fingers) without syndactyly in the left foot. There was a skin pit over the tibia (Figure 1). Radiography showed that the left fibula was absent in addition to the campomelic left tibia (Figure 2A), three metatarsals and phalanges in the left foot (Figure 2B). No additional pathological finding was

Figure 1. Picture showing the curved and short left leg, a skin pit on the tibia, and oligodactyly (3 fingers) in the left foot.

Figure 2A. Radiographs showing the absence of the left fibula, the campomelic left tibia.

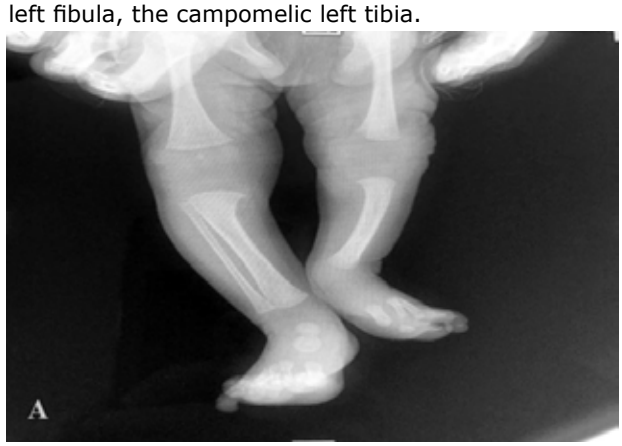
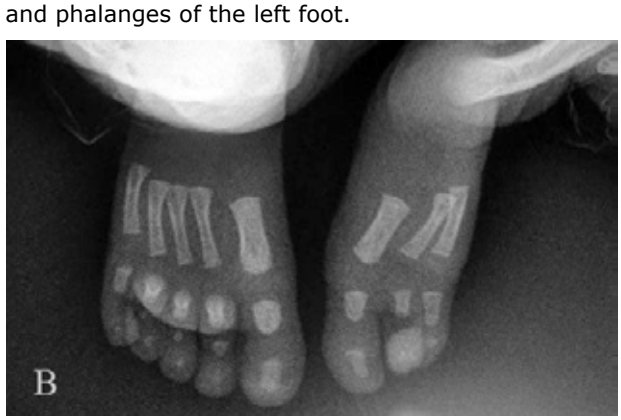


Figure 2B. Radiographs showing the three metatarsals and phalanges of the left foot.



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found on echocardiography, and cranial and abdominal ultrasonography. She was found to have normal 46, XX karyotype. After the parents were informed about the future limb lengthening surgery and orthosis, the patient was discharged for long-term follow-up.

What is the diagnosis?

Her physical examination and radiological findings were indicative of FATCO syndrome. Fibular hemimelia is a rare congenital limb anomaly characterized by partial or complete absence of the fibula with a spectrum ranging from mild fibular hypoplasia to complete fibular aplasia combined with malformation of the tibia and/or parts of the foot.[1] Fibular hemimelia or the absence of fibula is a rare defect with an incidence of 5.7 to 20 per million, and the exact etiology is unknown.[1] FATCO syndrome (MIM#246570) characterized by fibular aplasia, tibial campomelia and oligosyndactyly, first described by Hecht and Scott in 1981, is an extremely rare syndrome.[2] Although the syndrome is thought to be sporadic, it is also suggested that it has an autosomal dominant or X-linked inheritance.[3] To the best of our knowledge, this is the 24th case reported in the literature so far. Neurodevelopment has been reported to be normal in previous cases. Craniofacial anomalies reported in published cases included micrognathia, hypertelorism and hydrocephalus.[4] In addition, ventricular septal defect was reported in two of the cases.[5] Eight of 23 patients with FATCO syndrome diagnosed so far had also upper extremity anomalies. The upper extremity anomaly of six patients was bilateral and oligosyndactyly, which is the major feature of the syndrome, was frequently seen. As additional anomalies, the absence of radius and ulna was reported by Pentony et al.[6], and hemimelia in the left arm was reported by Heacht and Scott.[2] However, no additional anomaly was found in our patient.

In conclusion, fibular aplasia and tibial campomelia, which are the major components of FATCO syndrome, have been reported in all patients diagnosed so far. Oligosyndactyly in the hands or feet is seen in most cases. Although the pattern of inheritance is not certain, autosomal dominant inheritance with reduced penetrance is more likely. When counseling parents, it should be told that these children will show a normal neurodevelopment.

Compliance with Ethical Standards

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