

Fibular aplasia, tibial campomelia, oligosyndactyly (FATCO) syndrome: A rare disease with an evolving definition

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Abstract

Fibular aplasia, tibial campomelia, and oligosyndactyly (FATCO syndrome) is a rare disorder (estimated incidence of 5.7 to 20 cases/1 million births). Genetic basis and inheritance are unknown. Each case should be reported due to the paucity of reports and lack of a standardized treatment approach. To date, 15 cases of FATCO syndrome have been reported. We report two cases which match the radiological description of a FATCO syndrome variant and are the third and fourth reports on this variant. A 32-year-old female, G5/P2022 presented for her fetal anatomic survey at 20 2/7 weeks which showed a short left femur, short bowed left tibia, fibular hypoplasia and a left foot shorter than the right, leading to a diagnosis of FATCO (Fibular aplasia, tibial campomelia, oligosyndactyly) syndrome. She met with Pediatric Orthopedic surgery prior to delivery as a result of this finding. She will be followed throughout the rest of the pregnancy with serial growth ultrasounds. A 34-year-old female, G1/P0 was seen for the routine fetal anatomic survey at 21 3/7 weeks which showed a shortened/bowed right tibia and shortened right fibula. This led to the diagnosis of FATCO syndrome. She met with Pediatric Orthopedic surgery prior to delivery as a result of this finding and the rest of her pregnancy was uneventful with the spontaneous vaginal delivery of a baby boy at 40 weeks 1/7 days. FATCO syndrome ultimately requires corrective surgery after delivery. Prenatal identification of this syndrome allows for informed family planning. A detailed discussion with the pediatric orthopedic surgery as to the surgical timeline allows the mother and family to make informed decisions and plan for the necessary adjustments to their home life.



Biography

Jason Wheatley being educated in CPR and first aid in addition to lifesaving techniques from his days as a lifeguard to helping customers learn about their medications and insurance options as a pharmacy clerk. Dr Wheatley has his roots firmly in the science of healing. He is currently under training as a Maternal Fetal Medicine Fellow.

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