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Syndactyly Type IV/Hexadactyly of Feet Associated With Unilateral Absence of the Tibia

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We report on a newborn girl with syndactyly type IV, hexadactyly of feet, and right tibial hemimelia. She has 5 other relatives with identical anomalies of the hands and feet transmitted as an autosomal dominant trait. Syndactyly type IV is rare as is absence of the tibia. We suggest the possibility that syndactyly type IV may be a more complex entity, including lower limb malformations, and transmitted as autosomal dominant with variable expressivity.

KEY WORDS: **tibial hemimelia, lower limb malformations, autosomal dominant trait**

INTRODUCTION

Congenital absence of the tibia is rare. Only about 370 cases have been reported. Recently, we observed a newborn girl with absence of the right tibia and the rare syndactyly type IV/hexadactyly of feet.

CLINICAL REPORT

A 2805 g girl was delivered by cesarean section at 37 weeks of gestation after an uneventful pregnancy. Her Apgar scores were 9 and 10 at one and 5 minutes, respectively. No medication was taken during pregnancy.

At birth, she had hexadactyly of both hands. The thumbs were biphalangeal. All 6 fingers were fused with total fusion of proximal and distal phalanges giving the hands a cup-shaped form. She also had 6 well-formed and unfused toes on both feet. The right lower limb had a flexion-contracture at the knee. The right foot was in a rigid varus position and in supination pointing toward the perineum. The left lower limb was of normal length. No other clinical abnormalities were detected.

Roentgenograms showed normal hips, absence of the right tibia with hypoplastic lower femoral epiphysis, and normal right fibula. No abnormality of the left leg was noted. Both hands showed 6 metacarpals without synostosis. Corresponding phalanges were present without fusion. Both feet presented 6 metatarsal bones with their corresponding phalanges. Other skeletal bones were normal. Ultrasonography images of brain, kidneys, and heart were normal.

FAMILY HISTORY

Parents were unrelated Palestinian Moslems. The pedigree was traced over several generations. Only 3 informative generations are presented in Figure 1. The paternal grandmother has the same syndactyly type IV/hexadactyly of the feet. She is of normal intelligence and well adapted to everyday life although she did not have her hands repaired surgically.

The father and his sister had the same malformation as did 2 other sibs of the proposita. All were operated on for hands. These 5 members of the family were all examined by us and present exactly the same involvement of hands and feet as the proposita. None of them had absence of the tibia. No other cases of syndactyly, tibial hemimelia, or any other bone malformations were reported in the family.

DISCUSSION

This infant had type IV syndactyly, hexadactyly of both feet, and absence of the right tibia with hypoplastic lower femoral epiphysis described by Jones et al. [1978] as type I a congenital absence of the tibia.

Syndactyly type IV is a rare malformation. Only 3 cases, a mother and her 2 children, were reported by Haas [1940]. The syndactyly was complete, affecting the fingers of both hands with 6 metacarpals and 6 digits, and was associated with flexion of the fingers, giving the hands a cup-shaped form, without bone fusion. There was no mention of the condition of the feet and there were no associated malformations. The presence in the above described family of identical syndactyly in 5 other relatives over 3 generations, involving males and females, suggests autosomal dominant inheritance.

The incidence of tibial aplasia in the United States is

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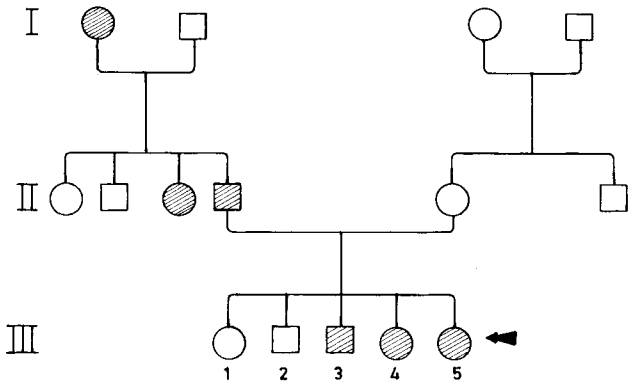

Fig. 1. Family pedigree.

approximately one per million live births and only about 370 cases have been reported [Wolfang, 1984]. Absence of the tibia has been described as an isolated malformation [Mc Kay et al., 1984] or in association with other abnormalities. Ferguson and Scott [1952] found that most cases were associated with anomalies such as dislocated hip, polydactyly, cleft palate, and imperforate anus.

In a study of 21 children, Kalamchi and Dawe [1985] found that most associated anomalies involved the hands (ectrodactyly, anomalies of the thumb), the testes (cryptorchidism, varicocele), the feet (severe equino varus deformity), the ipsilateral femur (hypoplasia with mild shortness), less frequent anomalies included hernia, learning disabilities, and scoliosis. Pratt [1971] reported 2 cases of lethal congenital cardiac defects associated with tibial aplasia.

Majewski et al. [1985] described 6 families with a total of 34 affected persons with the syndrome of tibial aplasia and ectrodactyly transmitted as autosomal dominant. Pashayan et al. [1971] reported bilateral aplasia of the tibia, polydactyly, and absent thumb in father and daughter, suggesting also an autosomal dominant inheritance. Cases of tibial aplasia associated with syndactyly have been described. Karamitsos and Bartsocas [1982] reported on an affected girl demonstrating hexadactyly on the right hand (radiologically, 6 metacarpals on each hand), bilateral cutaneous syndactyly of all fingers, absence of the distal 2/3 of both tibiae, and hexadactyly on each foot while her father presented polydactyly and syndactyly of both hands and partial syndactyly of the toes. Al-Awadi et al. [1982] reported on a large family in which the index patient showed, aside from hypoplastic bowed tibia, polysyndactyly of hands and feet while 16 relatives of both sexes in 4 generations only had either bilateral syndactyly or polydactyly or both. The polysyndactyly of the paternal grandmother was the first case reported in her family. This probable mutation was transmitted unmodified over 3 generations.

Could this syndrome, syndactyly type IV with hexadactyly of feet, be a more complex entity which includes a variety of lower limb malformations and inherited as autosomal dominant.

Though there are differences from family to family, these observations point to one (or several) autosomal dominantly mutant gene(s) causing limb malformations with considerably variable expressivity.

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