

TIBIAL APLASIA WITH ECTRODACTYLY - A RARE CASE REPORT

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ABSTRACT

BACKGROUND

Congenital absence of lower limb bones is very rare. Amongst the lower limb bones, tibial aplasia and hypoplasia being relatively common. Here, we report a sporadic rare case of tibial aplasia with ectrodactyly (Split hand).

KEYWORDS

Tibial aplasia, Ectrodactyly, Split hand, Cleft hand, Lobster claw hand.

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BACKGROUND

Tibial aplasia is an uncommon lower limb malformation that can occur isolated or be part of a more complex malformation pattern. Congenital absence of tibia is a rare and severe lower limb malformation with an incidence of approximately 1:1,000,000 live births.¹ Anomalies like Tibial hemimelia with split hand/foot malformation (TH-SHFM) and Gollop-Wolfgang complex are rare malformations with highly variable manifestations. The first case with this pattern of malformations was reported by Sir Ambroise Pare in 1575. In the present context, we report a rare case of Tibial aplasia with ectrodactyly with characteristic clinical and radiological features.

CASE REPORT

Here, we present a case born of a consanguineous marriage in Indian Hindu family. The patient presented with complaints of limping with deformed left leg and right hand. On clinical examination, child had malrotated thin left lower limb and absence of middle finger of right hand with stunted growth (Fig. 1). Skeletal survey of the child revealed aplasia of left tibia with hypoplastic 1st metatarsal bone on right side (Fig. 2) and radiograph of right hand revealed absence of middle finger of right hand giving a characteristic deep V shaped central bony defect (Split hand, cleft hand or lobster claw hand) - ectrodactyly (Fig. 3). Rest of the axial and appendicular skeleton was unremarkable. No positive family history of any genetic dysplasia. Prenatal period was uneventful, no documented antenatal scans was available.



Figure 1. Photograph of Child showing Aplasia of Left Tibia and Rudimentary Right First Metatarsal (Red Arrow)



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Figure 2. Radiograph of Both the Legs revealed Aplasia of Left Tibia with Hypoplastic 1st Metatarsal Bone on Right Side



Figure 3. Radiograph of Both Hands revealed Absence of Middle Finger of Right Hand giving a Characteristic Deep V Shaped Central Bony Defect (Split Hand, Cleft Hand or Lobster Claw Hand) - Ectrodactyly

DISCUSSION

Tibial aplasia-ectrodactyly syndrome is a rare condition characterised by congenital ectrodactylous limb malformations associated with tibial aplasia or hypoplasia. The incidence is estimated to be approximately 1 in 1,000,000 live births.¹ Majority of tibial defects are unilateral and sporadic.

The first case with this pattern of malformations was reported by Sir Ambroise Pare in 1575.² The first familial inheritance of this disorder was published by White and Baker in 1888.³

Limb anomalies are associated with number of factors like amniotic band syndrome, maternal exposure to antiepileptics and amniotic sampling, etc. They may also be inherited as an autosomal dominant manner with reduced penetrance. Duplication of BHLHA9 located on chromosome 17p13.3 is responsible for most of the limb anomalies. BHLHA9 is a basic loop-helix transcription factor.³

The Expression of the Phenotype is Highly Variable with Various Abnormalities as follows -

Limbs

Absence of long bones of legs in 55%, most commonly tibial aplasia/hypoplasia or fibular aplasia/hypoplasia.

Hand

Abnormalities in 68% of the affected. Most commonly ectrodactyly (Split hand) and absent of digits.

Feet

Abnormalities in 64% of the affected. Most commonly variable absence of tarsal, metatarsal and toes.

Occasional Abnormalities

Cup shaped ears, aplasia of ulna, radius or humerus. Syndactyly, bifurcation of femora, absence of multiple fingers, proximally placed thumb, postaxial and intermediate

polydactyly, talipes, supernumerary digits, patellar hypoplasia and hypoplasia of great toe.

Overlap with the Gollop-Wolfgang syndrome has been described. The syndrome is generally inherited in an autosomal dominant manner with reduced penetrance.¹ Autosomal recessive inheritance has also been proposed in some families. Two susceptibility loci at 1q42.2-q43 and 6q14.1 have been identified, leading to the hypothesis that this syndrome fits the model of digenic inheritance.

Jones et al classified tibial hemimelia based on initial radiographs.¹ Four types of deformity were recognised.

- Type I - Tibia is not visible.
- IA - Tibia is completely absent with a hypoplastic distal femoral end.
- Ib-A rudimentary tibia articulates with a relatively normal distal femoral end.
- Type II - Proximal tibia is preserved with a short tibial segment, but the distal tibial end is absent.
- Type III - Proximal tibia is absent.
- Type IV - Short tibia with distal tibiofibular diastasis is present.

In patients with a congenital absence of the tibia, accurate diagnosis is of the utmost importance in planning future treatment. In the absence of proximal tibial anlage, especially in patients with femoral bifurcation, the knee should be disarticulated.

Tibiofibular synostosis is a good choice in the presence of a proximal tibial anlage and good quadriceps function. The treatment of femoral bifurcation is simple - to resect at its base. However, the treatment of tibial hemimelia is challenging. Amputation, fibular transfer or reconstruction procedures are the alternative treatment options.¹

CONCLUSION

Tibial aplasia-ectrodactyly syndrome has variable expression with low penetrance of the gene responsible for this syndrome leading to normal phenotype even in presence of abnormal genotype. Hence, prenatal ultrasound should be performed in all pregnancies in affected families even if the parents do not manifest the syndrome clinically.

Familiarity of clinico-radiological features of this syndrome is essential postnatally for proper planning of future treatment with replacement or reconstruction.

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