



A Case of Bilateral Tibial Hemimelia Type IV and V with Bilateral Subdural Hygroma and Atrial Septal Defect

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ABSTRACT

Congenital absence of tibia is a rare anomaly, and may be total or partial, unilateral or bilateral. Total absence is more frequent than partial, unilateral absence occurs more often than bilateral, with right limb more commonly affected than the left. In partial defect, almost always the distal end of the bone is affected and, of the bilateral cases, there may be total absence on both sides, or total on one side and partial on the other. Males are slightly more commonly affected than the females. Though, the family history is usually negative for congenital abnormalities and other diseases, there is a considerable chance of occurrence of congenital defect of the tibia or of other abnormalities, in near or remote relatives. We report a case of newborn having bilateral tibial hemimelia type V (right) and IV (left).

Keywords: Aplasia, Congenital anomaly, Tibial defect, Tibial hemimelia.

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INTRODUCTION

Tibial hemimelia is a very rare disease with an incidence of 1 in 1,000,000 live births.¹ It is characterized by deficiency of tibia with relatively intact fibula. It causes marked shortening of the involved extremity with a severe equinovarus deformity.² The defect could be either complete or incomplete, and occurs either as a solitary disorder, or as a part of more complex malformation syndrome.^{1,3} Associated abnormalities include those of the musculoskeletal system of both lower and upper limbs, orofacial part, urogenital and cardiovascular systems. The tibial reduction defects were initially classified into four major types based only on X-ray

findings,⁴ however, later expanded to seven major types and five subgroups according to the therapeutic relevance and considering the importance of cartilaginous anlage (Table 1).⁵

According to the Weber classification, the sequence of the distribution of tibial hemimelia types is as follows: 62% of cases with type VII, 15% with type III, 6% with type I, 6% with type V, 5% with type II, 3% with type IV, and 3% with type VI. If the older classification corresponds to Weber classification, then the sequence of the distribution would have been as follows: 61% of cases with type I, 16% with type II, 5% with type IV, 3% with type III, and 15% with not otherwise specified. We describe a rare case of bilateral tibial hemimelia that would be classified as type I in older classification, however, type VII according to the Weber classification.

CASE REPORT

A male infant was born at 37 weeks gestation to a 24-year-old primigravida. The antenatal period was mostly uncomplicated, without any history of diabetes or hypertension. His mother was a nonsmoker. There was no history of any teratogenic drug intake by the mother including routine multivitamin supplementation also. There was history of febrile illness in second month of pregnancy.

Antenatal ultrasound carried out in another hospital showed normal amount of amniotic fluid. Abnormalities, all located in the bilateral lower limbs included, club foot, tibial agenesis, and distal femoral bifurcation. The upper limbs were normal, and there were no other notable malformations. Parents were of Indian descent with no evidence of consanguinity.

The neonate was born by normal vaginal delivery. His birth weight was 2.6 kg (appropriate for gestational age), head circumference was 33.5 cm (50-75th centile), and upper segment length was 29 cm (normal). He did not require any intervention at birth.

On examination, both knees were flexed and held at 30°. Both feet showed equinovarus deformity. The leg segment was internally rotated to 60° bilaterally and bony prominence was seen on the lateral aspect of the knee joint on both sides (Fig. 1). The child had an absent quadriceps mechanism with dislocated dysfunctional knee and ankle joints bilaterally associated bilateral feet

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Table 1: Classification of tibial hemimelia

Newer (Weber) classification ¹		Older classification (Jone's classification)	
I	Tibial hypoplasia	I ^a	Tibia not seen. Hypoplastic lower femoral epiphysis
II	Tibial diastasis	I ^b	Tibia not seen. Normal lower femoral epiphysis
III (III ^a , III ^b)	Distal tibial aplasia	II	Distal tibia not seen
IV (IV ^a , IV ^b)	Proximal tibial aplasia	III	Proximal tibia not seen
V (V ^a , V ^b)	Bifocal tibial aplasia	IV	Diastasis (both proximal and distal epiphyses are present, but the tibia is short)
VI (VI ^a , VI ^b)	Tibial agenesis with double fibulae		
VII (VII ^a , VII ^b)	Tibial agenesis with single fibula		



Fig. 1: Bilateral lower limb deformity



Fig. 2: Bilateral foot deformity



Fig. 3: X-ray showing tibial deformity

deformity (Fig. 2). The radiological evaluation revealed a normal hip joint. The lower end of femur was normal and patella was present. Complete absence on left and partial on right was noted with small cartilaginous anlage, and fibula was present on both legs without any sign of doubling (Fig. 3). Calcaneus was displaced anteriorly and was higher than the lower end of fibula. Both the right

and left foot had three tarsal bones, four metatarsals, and three toes each having two phalanges each on left (Fig. 4). The musculoskeletal system of upper limb did not reveal any abnormality, and there was no dysmorphic facies. Systemic examination also did not reveal any abnormality, except that the genitalia were dark pigmented. The newborn was started on breast-feeding, which he was accepting well. Magnetic resonance imaging of brain was suggestive of thin symmetrical hygroma in bilateral frontotemporoparietal region. Doppler study of both lower limbs was normal arterial and venous blood flow. Two-dimensional echo and color Doppler was suggestive of patent foramen ovale about 3 mm.

DISCUSSION

The limb defects appear to have a multifactorial etiology, arising from an interaction between environmental influences, teratogenic exposure and the individual's genetic makeup. Tibial hemimelia syndrome, which is a rare autosomal dominant condition, encompasses several types of syndrome, all having a common phenotype of tibial hypoplasia or agenesis and polydactyly.⁶ Family history of congenital anomaly of tibia as well as intrafamilial phenotypic variations of the defect in twin pregnancy has been reported.⁷⁻⁹ The anomalies in which the number of skeletal elements increased arise during the first 7 weeks

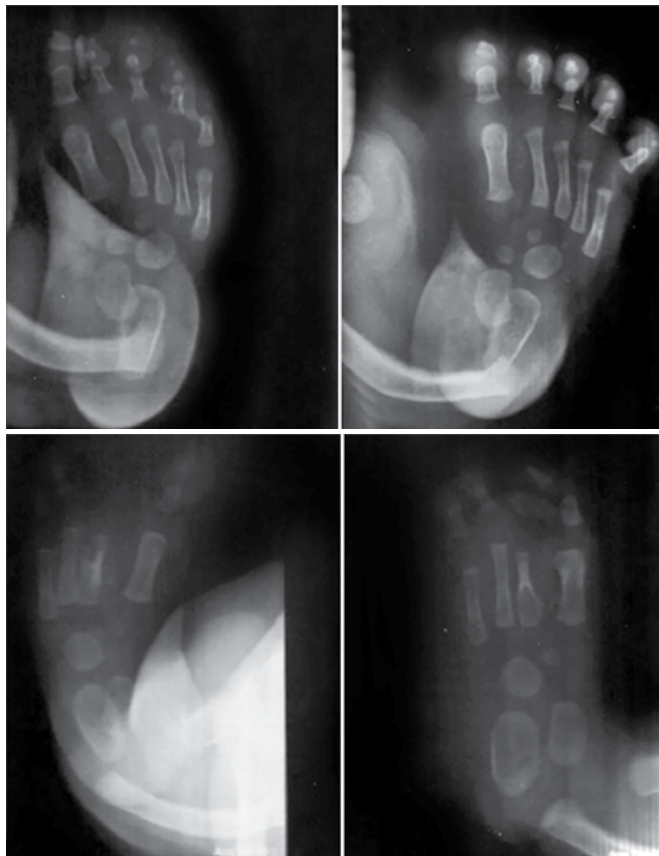


Fig. 4: X-rays showing bilateral feet deformity

of intrauterine life. A decrease in the number of skeletal parts may arise after as well as during, this 7 weeks period.¹⁰ The mother of the index case was suffering from pyrexia of unknown origin during the first trimester, and this might have led to the defect by its interaction with some unidentified environmental influences and absent intake of multivitamin supplementation by the mother.¹¹

As described above, tibial hemimelia is usually accompanied by other congenital anomalies that include; congenital dislocation of the hip, ectro-, poly-, or syndactyly, abnormalities of the musculoskeletal system of both lower and upper limbs, phocomelia, harelip and cleft palate, pseudo-hermaphroditism, cryptorchidism, and hypospadias, etc. One report described a case of tibial hemimelia, femoral bifurcation, cleftlip/palate, and cardiovascular anomaly like atrioventricular canal with truncus arteriosus.¹² However, in the index case, though there was bilateral tibial hemimelia with foot ectrodactyly, systemic malformations were absent except for the hyperpigmented genitalia, patent foramen ovale and cystic hygroma in brain.

Some cases of tibial hemimelia are genetically transmitted, whereas others are sporadic. Few syndromes, such as tibial hemimelia, foot polydactyly triphalangeal thumbs syndrome (Werner syndrome), tibial hemimelia diplopodia syndrome, tibial hemimelia-split hand/foot

syndrome, tibial hemimelia-micromelia-trigonbrachycephaly syndrome, tibial hemimelia-normal upper limb syndrome and tibial hemimelia-radial agenesis syndrome, are considered to be transmitted as autosomal dominant. In others, such as tibial hemimelia-cleftlip/palate syndrome, tibial hemimelia split hand/foot syndrome, an autosomal recessive inheritance is suggested.¹³ In some situations, a cystic hygroma can be present in a healthy baby. If a chromosome abnormality is not found in the fetus, the outcome is generally better than for those who do have a chromosome abnormality. Overall, there is generally a poor prognosis associated with the prenatal finding of cystic hygroma. Cystic hygromas that develop in the third trimester (after 30 weeks gestation) or in the postnatal period are usually not associated with chromosome abnormalities.¹⁴

Treatment mainly includes surgical correction of the deformity where possible. Common surgical procedures include disarticulation at knee, Syme's amputation or Chopart's amputation.^{2,7} The sooner the amputation is performed the easier and faster the rehabilitation and adaptation to the prosthesis. The absence of the cartilaginous anlage increases the difficulty of the operative procedure. The surgical option for the index case with favorable result would include disarticulation of the knee joint and prosthesis.²

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