

# Case report: Congenital absence of tibia - Type 2

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## Abstract

Congenital tibial absence is a rare anomaly causing shortening and varus deformity of the lower extremity. It may present as an isolated anomaly or be associated with a variety of skeletal and extra skeletal malformations. We describe a newborn baby with congenital absence of right tibia (type 2) and Patent ductus arteriosus.

Key words: Absent tibia, Congenital anomaly, Limb defect  
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## Introduction

Congenital absence of tibia is one of the rarest of the congenital malformations, and certainly the most uncommon of those of the lower limb (1). Only few cases have been described in Indian literature and hence the report.

A term female baby born by caesarean section for trans-verse lie was brought to NICU with deformity of right lower limb. The parents were non consanguineous, the antenatal period was normal with no exposure to teratogenic drugs or radiation. Their previous two babies (both females) had died in early infancy.



Figure 1. Right lower limb deformity with the bony (tibial) rudiment



Figure 2. X-ray of the right lower limb showing deficiency of tibia

The exact cause of their deaths was not known but they apparently had no malformations. On examination, her cry, colour and activity were good. Her right lower limb was shortened and deformed. The right leg was flexed and adducted and the ipsilateral foot had a varus deformity. A bony rudiment was noted in the upper medial aspect of the leg (Fig.1). Only a single bone (fibula) was palpable and the lateral malleolus was prominent. The other limbs including the digits were normal. Systemic examination did not reveal any abnormality. X-ray of the right lower limb showed deficiency of tibia and a thick curved fibula.

A small portion of the proximal tibia was noted corresponding to the rudiment palpated (Fig.2). Echocardiography revealed a Patent ductus arteriosus with left to right shunt and hemogram showed a normal platelet count. The parents have been advised follow up for both the ductus and the limb deformity.

## Discussion

Congenital absence of the tibia is defined as “the absence at birth of the tibia in whole or in part, the lower limb being nevertheless present with its different segments well developed in their essential parts.” It may be total or partial, unilateral or bilateral. Total absence is more frequent than partial, unilateral absence occurs more often than bilateral, and of the unilateral cases the defect is of the right limb more frequently than of the left. A noteworthy feature of the partial cases is that the defect is almost always of the distal end of the bone. With regard to sex there is a slight preponderance of males over females [1].

It may present as an isolated anomaly or be associated with a variety of skeletal and extraskelatal malformations. Tibial hemimelia may also constitute a part of a more complicated malformation complex or syndrome, such as the Gollop-Wolfgang complex and triphalangeal thumb-polysyndactyly syndrome [2,3]. In this baby, patent ductus arteriosus is an associated malformation and more lethal cardiovascular anomalies have been reported earlier [4]. The three features namely shortening of the leg, flexion and adduction at the knee, and marked varus of the foot point towards the diagnosis and can be confirmed by an X-ray. Prenatal diagnosis using ultrasound is also possible. Jones et al. classified the anomaly into 4 types according to radiologic criteria and the anomaly of this baby fits in type 2 where the proximal epiphysis is preserved with a short segment of tibia [5]. Correct interpretation of radiological signs at an early age is essential to rational management. In particular, we must avoid an unnecessarily mutilating amputation through the knee when there is a prospect of preserving the joint in those in whom the tibia, although

translucent, is not totally absent. Bombaci et al have described a five-year-old boy with bilateral congenital tibial deficiency of type 2 who was treated with tibia-fibular fusion without Syme amputation [6].

The etiology for this anomaly remains uncertain. The germinal theory regards a primary defect of the mesoblast formation from which the skeleton of the affected limb is developed. Tibial development defect in valproic acid embryopathy has also been reported [7]. However, in this baby, no specific cause for the anomaly could be found. Reconstructive surgery and a prosthesis adapted to growth, together with regular post-operative follow-up, are necessary for optimal functional results.

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