CONGENITAL AGENESIS OF THE TIBIA ASSOCIATED WITH ABSENCE OF THE THUMB

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ABSTRACT

A 1½ year old boy with congenital agenesis of the tibia associated with absent thumb and equinovarus deformity on the same limb was presented.

Key words: Agenesis, tibia, thumb.

INTRODUCTION

Complete congenital longitudinal deficiency of tibia is an extremely rare disorder with unknown etiology that results in shortening and equinovarus deformity of the foot. The incidence of the disease is about one per million (1).

CASE REPORT

A 1½ year old boy was admitted with the deformity on the right lower extremity. He was born as a second child of nonconsanguineous parents. No such deformity was described in both side relatives. Physical examination revealed a well child. His right lower extremity showed marked inversion and adduction with flexion contracture on the knee and proximal displacement of the fibular head. He had also oligodactyly (four toes) on the same limb (Figure 1). No other associated anomaly was detected. Routine laboratory examinations, EKG, abdominal ultrasonography and chest x-ray were within normal limits. Complete absence of the right tibia was observed on the radiographs (Figure 2, 3).

DISCUSSION

Complete congenital longitudinal deficiency of tibia is an extremely rare disorder with incidence of about one per million (1).
Figure 1. Oligodactyly on the limb

Figure 2, 3. Radiographs showing absence of the tibia
Although some authors suggest that abnormality of vessels including reduction in number and extravasation of blood or embolisation on the limb in embryonic period lead to absence of tibia, the etiology still remains unknown (2). The disease is classified by Kalamchi and Dawe (3) into three types: In type I, the tibia is totally absent, in type II, the distal half of the tibia is absent, in type III, distal deficiency of the tibia is associated with tibiofibular diastasis. Our patient is obviously in type I according to this classification. Kalamchi and Dawe (3) also reported that two thirds of the cases have also associated anomalies. These include syndactyly, polydactyly, absence of thumbs or bifid thumbs, equinovarus deformity, femur deformity, congenital heart disease, absence of the radius, duplication of the ulna and fibula, hernia, cryptorchidism and varicocele (4-6). In our case none of these was existed but oligodactyly and equinovarus deformity.

In the treatment of the deformity surgical intervention particularly fibular transfer is needed (7).

REFERENCES


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