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An infant with congenital fibula deficiency accompanying with deafness

F. CEKMEZ, O. PIRGON, F.E. CANPOLAT

Department of Pediatrics, Gülhane Military Medical Academy, Istanbul, Turkey

Abstract. – Congenital longitudinal deficiency of the fibula (CLDF) is the most common congenital defect involving the long bones. There have been many different classifications developed for fibula deficiency. Achterman and Kalamchi's classification is most commonly used and will be described. Our case was complied with type II. Other anomalies includes cardiac anomalies, thrombocytopenia absent-radius (TAR) syndrome, thoracoabdominal schisis, spina bifida and renal anomalies, but most associated anomalies are skeletal. We here presented firstly in literature an infant with congenital fibula deficiency accompanying with deafness.

Key Words:
Fibula deficiency, Deafness.

Introduction

Congenital longitudinal deficiency of the fibula (CLDF) is the most common congenital defect involving the long bones. Males are affected twice as much as females. The precise etiology remains unknown and thought to be secondary to an insult that occurs to the limb bud during the second to eighth week of fetal development. Genetic factors do not appear to be involved, certain environmental factors (i.e. insulin, irradiation) are capable of producing this deformity[1]. We here presented firstly an infant with congenital fibula deficiency accompanying with deafness in literature.

Case Report

This is a 2 year old female who was the product of a NSVD at 38 weeks gestation without complications. Soon after birth the patient was noted to have a severe deformity involving the right leg and right foot. Radiographs at that time revealed absence of the fibula on the right with ipsilateral hypoplasia of the tibia. There is no family history of similar deformities and no identifiable environmental insult during pregnancy. Examination of the right lower extremity demonstrates shortening of the right thigh with a 10 degree flexion contracture present in the knee. The knee is noted to be in valgus. Overall, the right lower extremity is noted to be 5 cm. shortened when compared to the contralateral normal extremity. In addition to these, she had deafness but other systems were normal.

Discussion

There have been many different classifications developed for fibula defieciency, but Achterman and Kalamchi's classification[1] is most commonly used and will be described.

Type IA – A portion of the fibula remains present. The proximal fibular epiphysis is distal to the level of the proximal tibial physis while the distal fibular physis is proximal to the talus.

Type IB – There is partial absence of the fibula (30-50%) with the distal remaining portion unable to function in ankle support.

Type II – Complete absence of the fibula.

This classification is useful as a prognostic indicator and for aiding the decision-making process regarding the treatment. Our case was complied with type II. Other anomalies include cardiac anomalies, thrombocytopenia absent-radius (TAR) syndrome, thoracoabdominal schisis, spina bifida and renal anomalies, but most associated anomalies are skeletal and includes[2-7] peromelia (severe anomalies of distal limbs including absence of hand or foot), congenital dislocation of the head of the radius, craniosynostosis, syndactyly, brachydactyly and clinodactyly of fingers and toes, onycho-osteodysplasia[8], facial dysmorphism, congenital shortening of the femur, distal femoral valgus with condylar hy-

Corresponding Author: Ferhat Cekmez, MD; e-mail: ferhat_cocuk@hotmail.com

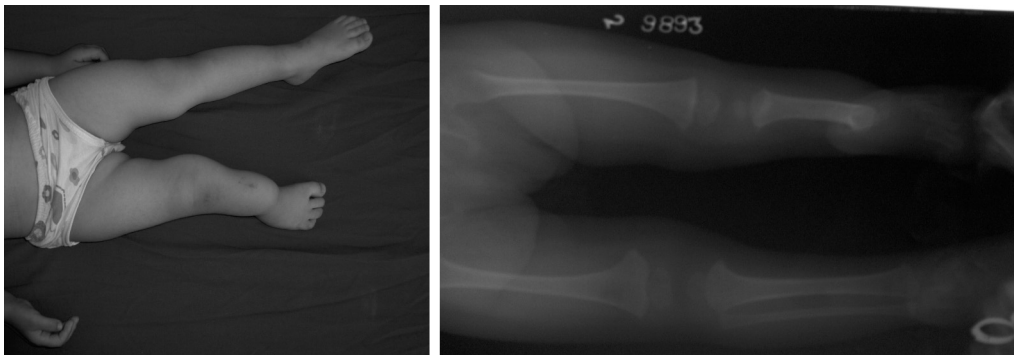

Figure 1. ***A, B,*** Absence of the fibula on the right leg with ipsilateral hypoplasia of the tibia.

poplasia, external rotation deformity. Up to now there is no any report about fibula deficiency accompanying with deafness; because of this reason we tried to inform this togetherness.

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