

Congenital short femur: A case report

Konjenital kısa femur: Olgu sunumu

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Abstract

Congenital short femur (CSF) is a very rare condition and after the ultrasonography has been actively used, the placement of the antenatal diagnosis is actually quite facilitated. It is usually not accompanied by chromosomal abnormalities. Our case presentation has shown the importance of both femur lengths and all other extremity images while performing antenatal scanning.

Keywords: Congenital short femur, Proximal focal femoral deficiency, Antenatal diagnosis

Öz

Konjenital kısa femur çok nadir görülen bir durumdur ve ultrasonografinin kullanılması sonrası antenatal olarak teşhis edilmesi oldukça kolaylaşmıştır. Genellikle kromozomal anomaliler eşlik etmez. Bizim vaka takdimimiz ultrasonografik tarama yaparken heriki ekstremitte uzunluklarının ölçülmesinin hatta bütün ekstremitte uzunluklarının ayrı ayrı ölçülmesinin önemini vurgulamaktadır.

Anahtar kelimeler: Konjenital kısa femur, Proximal fokal femoral deficit, Antenatal tarama

Introduction

Congenital short femur (CSF) is an extremely rare limb anomaly with an incidence of 1.1–2 in 100 000 live births. The diagnosis of this anomaly has been enhanced by widespread use of ultrasonography [1]. CSF is not usually accompanied with chromosomal abnormalities and mental disorders. The results of surgical repair are usually good [2]. Determine whether isolated femoral abnormality is part of the syndrome is the most important issue [3]. However, some congenital short femur cases involving skeletal disorders may be accompanied by global dysplasia syndromes and termination of gestation may be a reasonable option in such cases [4].

The proximal end of the femur is congenital absent. Most cases have been reported in the orthopedic literature and in the radiological literature. Femur defects are highly heterogeneous, and both femur length and proximal shape change significantly. Although a familial event has been reported, the mode of transmission is unknown [5]. CSF is frequently mixed with Unilateral Isolated Proximal Femoral Focal Deficiency (PFFD). But these two anomalies are different. CSF is the isolated shortness of the femur. Unilateral Isolated Proximal Femoral Focal Deficiency is an entity that includes the proximal absence of the femur from the partial absence of the femur. These two situations are actually different. Several classification schemes for PFFD have been proposed. This classification is between the acetabulum and the proximal end femur anatomical relationship accounts and has prognostic significance. The classification of Aitken is based on radiographic view [6]. The disease varies from a benign form (A) to a severe form (D) according to the degree of femoral insufficiency. In our study, we report a case of isolated, unilateral congenital short femur detected in a postpartum patient.

Case presentation

A 27-year-old pregnant woman admitted to give birth. This pregnancy was the first gestation of the patient. We found unilateral short femur during routine baby examination after delivery (Figure 1). The family did not have a story about skeletal anomalies or other illnesses. There was no relationship between mother and father and both parents were healthy.

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No thoracic, cardiac, gastrointestinal, genitourinary and neurological anomalies were observed in fetal findings. There were no other skeletal anomalies. There was no history of the mother about diabetes, drug use, exposure to teratogenic radiation, and viral infection during pregnancy. It was routinely followed during the pregnancy and there was no high-risk condition in the screening tests.



Figure 1: X-ray of the baby

Discussion

CSF is usually sporadic, and about 85-90% of cases are unilateral. Despite the fact that the CSF has several familial reported cases, the genetic pathway is not known [6-7]. Prenatal diagnosis of femoral anomalies is possible. But only 19% of the cases are prenatal diagnosed and 68% are diagnosed postnatally [8].

The critical period for skeletal development is the first 4 and 8 weeks. As a result, exposures during these periods (such as poor diabetic control, thalidomide-like drug use narrative, viral infections, radiation, focal ischemia, chemical toxicity, trauma, familial transitions) may be risky [9,10,11]. Some cases diagnosed at approximately 14 weeks of gestation by transvaginal ultrasonography have been reported in the literature. Some cases diagnosed at approximately 14 weeks of gestation by transvaginal ultrasonography have been reported in the literature.

CSF is actually a femur dysplasia. But it is often mistakenly diagnosed as PFFD although PFFD is completely different in terms of radiological and functional outcomes [12].

Many diseases (kyphomelic dysplasia, campomelic dysplasia, osteogenesis imperfecta, achondroplasia, achondrogenesis, thanatophoric dysplasia, short limb polydactyly and malformations of skeletal dysplasia) are usually considered

in differential diagnosis, but these diseases often affect other bones [13].

Prenatal diagnosis is also very important because of the contribution of the developing technology. Ultrasonography scanning is sometimes performed with single extreme measurements for measurement. For this reason, these anomalies and single extremity development disorders can be overlooked.

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