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Case Report

A Case of Bilateral Agenesis of the Femur

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Abstract
Bilateral femoral agenesis is a rare anomaly. To the best of our knowledge, only three cases of simple congenital anomaly and three cases associated with femoral facial syndrome have been reported. Here, we describe a simple form of bilateral femoral agenesis observed in one of the 2 dead fetuses delivered after termination of a 24-week twin pregnancy of a normal mother. Post-mortem x-ray examination confirmed the agenesis of both femurs and also left fibula.
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Keywords • Femur • agenesis • congenital

Introduction

The embryonic limb bud is derived from numerous embryonic tissues including surface ectoderm, somatopleural mesoderm, neural crest cells, and somites.[1] Specialized regions of the developing limb bud such as the zone of polarizing activity and the apical ectodermal ridge, direct and coordinate the development of the limb bud.[2]

Researchers have proposed many molecular and cellular mechanisms controlling normal development of the limb structure.[3-5] It has been well-documented that genetic as well as environmental factors might interfere with these normal sequence of events and cause varieties of defects including femoral agenesis.

Case Report

A 34-year-old multi-gravida with twin pregnancy was admitted for caesarian delivery at her 24th week of gestation. The mother had no history of diabetes mellitus, and cigarette smoking. There was no consanguinity between the parents. A few days prior to admission, the mother had felt no fetal activity. Sonography confirmed fetal death. After delivery of the fetuses through a cesarian section, they were sent to the Pathology Department. Autopsy showed two female fetuses with one placenta. The larger fetus weighted 685 g with no abnormality or congenital defects. The smaller fetus weighted 415 g with a crown-rump length of 16 cm and a crown-heal length of 20 cm (ratio; 1/1.25, normal: 1/1.45). Post-mortem x-ray examination showed bilateral agenesis of femur and agenesis of left fibula (Fig 1). As seen in Fig. 1, the legs were directly connected to the body. We did not observe any other anomalies in the skeleton or internal organs. Histopathologic examination showed no significant abnormality.

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Discussion

Bilateral femoral agenesis is a rare and unusual anomaly and to the best of our knowledge, only six cases have so far been reported.[6-8] Three of these six cases were associated with femoral facial syndrome from diabetic mothers. This disorder is more common in females.[9-12] Although there is a strong relationship between femoral-facial syndrome and maternal diabetes, the etiology remains unknown.[11,13]

Human limb buds grow and differentiate between the 4th and 8th weeks of gestation.[4,5] Inadequate control of diabetes in the mother, as well as, viral infections, irradiation, focal ischemia, abdominal trauma, and drug exposure during this early critical phase may account for the subsequent developmental anomalies.[6,10,14] However, none of the above-mentioned factors could be identified in our case.

The etiology of the congenital femoral agenesis is unknown; however, autosomal dominant mode of inheritance is suggested in one case of one affected father and his daughter.[2] Many of the genetic syndromes are associated with congenital malformations, and 0.2% of newborns have severe limb anomaly.[2,6] Viral infection, drug, and radiation exposure during pregnancy are among environmental factors.[2] Less than 1% of congenital malformations are caused by drugs such as thalidomide, androgens, alcohol, and warfarin which are teratogenic agents.

To the best of our knowledge, this is the first case of simple bilateral congenital twin pregnancy femoral agenesis accompanied by left sided agenesis of fibula.

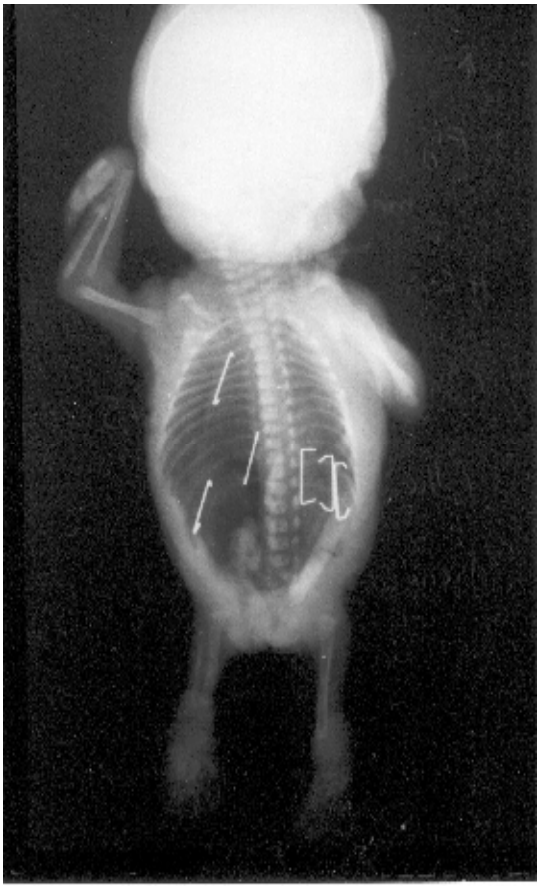

Figure 1: Bilateral congenital agenesis of the femur and left fibula

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