Bilateral femoral agenesis in 3-month-old baby: A case report

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INTRODUCTION

A complex cascade of growth factors controls fetal and lower limb development. These factors are expressed at different levels in a particular order during development. After fertilization, 4 to 8 weeks later, the extremities begin to develop. Most limb deformities manifest during this time, particularly during the fast proliferation and differentiation of cells and tissues, which peaks between 5 and 6 weeks after conception. Simple hypoplasia to complete absence of bone is all congenital abnormalities of the femur. For a guide to predicting limb development, it is crucial to make a clinical differentiation between various forms of femoral abnormalities.¹⁻³ We report this case because it is rare and needs proper evaluation and management. But since most cases are isolated, parents may rest comfortably that there is little chance that their subsequent children would also be impacted.

CASE REPORT

A 3-month-old baby girl presents with a complaint of short stature. She was born spontaneously at 38 weeks of gestation with a birth weight of 2575 g, a height of 36 cm, and a head circumference of 31 cm. The patient was born remarkably and cried soon after birth. The patient is the first child. There was no consanguinity between the parents. The mother has no history of chronic diseases such as hypertension and diabetes mellitus. During pregnancy, the patient’s mother had a routine antenatal care ultrasound examination twice by an obstetrician in Makassar, and no abnormalities were found. The patient's blood pressure and blood sugar levels during pregnancy were within normal limits. Drugs consumed during pregnancy are iron supplements and folic acid tablets. Both of the patient’s parents do not smoke. There was no history of exposure to other teratogens. No abnormalities were found in the father’s and mother’s families.

The patient’s condition is fit and fully breastfed since birth. There is no fever, cough or runny nose. Patients receive routine immunizations according to schedule. Physical examination showed the general condition was good, respiratory rate 42 x/minute, pulse 110 beats/minute, axillary temperature 36.7°C. In general status, the head was normocephalic; on examination, both eyes did not appear anemic and did not have jaundice. Examination of the ears, nose, and throat found no abnormalities and no enlarged lymph nodes in the neck. On examination of the thorax, the heart obtained a single heart sound (S1 and S2), regular, with no murmurs and gallops. In the lungs, breath sounds were vesicular, and no rhonchi or wheezing were found. On abdominal examination, bowel sounds were within normal limits; there was no distension, palpable tenderness, no liver and spleen enlargement, and a palpable mass. Upper and lower extremities were warm, dry

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Received: 2022-09-05
Accepted: 2022-10-01
Published: 2022-11-07

ABSTRACT

Background: Bilateral congenital femoral agenesis is a rare congenital anomaly. To our knowledge, there have only been three examples of simple congenital abnormalities linked to the femoral facial syndrome. We present a straightforward case of bilateral femoral agenesis in a 3-month-old female infant who did not have femoral facial syndrome and whose mother did not have diabetes.

Discussion: Bilateral femoral agenesis is a rare and unusual anomaly; only six cases have been reported. Other structural anomalies are often involved; these include the acetabulum, musculature, vessels, and ligaments of the knee, tibia, fibula, and foot. In our case, there are no other anomalies found. The majority of cases, however, are sporadic; therefore, the parents may be reassured that the risk of further offspring being affected is negligible. The classification by Aitken is widely used. This classification is based on the severity of the hip and femur radiographic findings.

Case Presentation: A 3-month-old female baby presents with a complaint of short stature. The baby was born full term spontaneous labor, 2570 grams vigorous and started to cry immediately after birth. The mother has no history of diabetes, cigarette smoking, and exposure to teratogens. The parents were not related by blood. X-ray examination showed bilateral agenesis of the femur, normal tibia and fibula, and proximal cruris in lateral acetabula.

Conclusion: Bilateral congenital femoral agenesis is a rare congenital anomaly. Although frequently related to maternal diabetes mellitus, there is no recognized cause for most cases.

Keywords: femoral agenesis, congenital deformity.


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Femoral hypoplasia is a rare but complex major limb defect, ranging from a simple shortening of the femur to complete femoral agenesis. Only six occurrences of bilateral femoral agenesis have been documented to date, making it a rare and unusual condition. In females, this disease is more prevalent. These anomalies can be isolated or co-occur with another anomaly, such as clubfoot, congenital heart problems, spinal dysplasia, congenital fibular hemimelia (most frequent), or a lack of lateral foot rays. Other structural abnormalities are frequently involved, such as those affecting the acetabulum, muscles, blood vessels, and ligaments in the knee, tibia, fibula, and foot. In our case, there are no other anomalies found.

The process of forming bones is called osteogenesis or bone ossification. This process, which varies for each person, starts during embryonic development’s sixth and seventh weeks and lasts until roughly age 25. There are two types of bone ossification, intramembranous and endochondral. A precursor of mesenchymal tissue is the starting point for each of these processes. The flat bones of the skull, clavicle and most of the skull’s bones are created by the direct conversion of mesenchymal tissue into bone, known as intramembranous ossification. Mesenchymal tissue first becomes cartilage during the process of endochondral ossification. Mature bone subsequently replaces the cartilage to build the axial skeleton and long bones.

Congenital femoral agenesis has an unclear origin, but in one affected father and his daughter, an autosomal dominant mode of inheritance has been proposed. Congenital deformities are linked by genetic diseases, and 0.2% of newborns have severe limb anomalies. The subsequent developmental defects may be related to the mother’s inadequate management of her diabetes, as well as viral infections, irradiation, localized ischemia, abdominal trauma, and medication exposure during this crucial early period. There was some of the axis femur associated with femoral facies syndrome. The most important characteristics are The femurs’ hypoplasia and a distinctive facial profile with a short nose, large philtrum, thin upper lip, and micrognathia.

DISCUSSION

Limb defects are relatively frequent congenital defects, but they occur mostly in the upper extremities and are, in most cases, minor – as polydactyly, supernumerary digits, syndactyly, or fusion of two or more digits. Congenital femoral hypoplasia is a rare but complex major limb defect, ranging from a simple shortening of the femur to complete femoral agenesis. Only six occurrences of bilateral femoral agenesis have been documented to date, making it a rare and unusual condition. In females, this disease is more prevalent. These anomalies can be isolated or co-occur with another anomaly, such as clubfoot, congenital heart problems, spinal dysplasia, congenital fibular hemimelia (most frequent), or a lack of lateral foot rays. Other structural abnormalities are frequently involved, such as those affecting the acetabulum, muscles, blood vessels, and ligaments in the knee, tibia, fibula, and foot. In our case, there are no other anomalies found.

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Nevertheless, none of the
aforementioned factors could be found in our circumstances. Other common features include Talipes equinovarus, syndactyly, preaxial polydactyly, cleft palate and lip, upslanting palpebral fissures, and microtia. Additionally related are disorders of the coccyx, spine, lungs, heart, kidneys, and urinary system.\textsuperscript{10-12}

Aitken's classification is frequently used. The severity of the radiographic findings in the hip and femur is the basis for this categorization. Aitken categorization has four categories: Although descriptive, Aitken categorization is not useful for treatment. Paley and Guardo created the classification system for congenital femur deficit based on the variables that affect lengthening repair. The matter at hand does not fit into any of the aforementioned categories.\textsuperscript{12,13}

**CONCLUSION**

An extreme case of congenital femur insufficiency is congenital femur absence. This anomaly can be present alone or in conjunction with another anomaly, such as clubfoot, congenital heart abnormalities, spinal dysplasia, or the most prevalent fibular hemimelia. There are many categories for this peculiarity, but none apply to our situation. The patient and her parents may benefit from additional assessment and management of this situation to assist the patient in accepting her condition and leading a satisfying social and economically productive life.

**CONFLICT OF INTEREST**

We declare that there were no conflicts of interest in this study.

**FUNDING**

The authors are responsible for the study funding without the grant, scholarship, or other funding resources.

**AUTHOR CONTRIBUTION**

All of the authors equally contributed to the study.

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