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Bilateral Femoral Agenesis In 3-Month-Old Baby: A Case Report

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Abstract

Background: Bilateral congenital femoral agenesis is a rare congenital anomaly. To the best of our knowledge, only three cases of simple congenital anomaly, three cases associated with femoral facial syndrome, have been reported. We describe a simple form of bilateral femoral agenesis observed in 3-month-old female baby without femoral facial syndrome and non-diabetic mother.

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

Discussion: Many of the genetic syndromes are associated with congenital malformations, and 0.2% of new-borns have severe limb anomaly. 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. There was some of agenesis femur associated with femoral facies syndrome. Most relevant features include hypoplasia of the femurs and a faces with short nose, long philtrum, thin upper lip and micrognathia. Other frequent findings are talipes equinovarus, syndactyly and preaxial polydactyly, cleft palate and/or lip, upslanting palpebral fissures and microtia. Costal, vertebral, lung, heart, renal and urinary abnormalities are also associated

Conclusion: Bilateral congenital femoral agenesis is a rare congenital anomaly. Although commonly associated with maternal diabetes mellitus, most cases have no known cause.

Keywords: femoral agenesis, congenital deformity

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Introduction

Fetal growth and lower limb development are controlled by a complex cascade of various growth factors. These factors are expressed at different levels in a particular order during development. Embryogenesis in the extremities occurs 4-8 weeks after fertilization. Most limb defects occur during this period, especially during rapid proliferation and differentiation of cells and tissues, peaking at 5 and 6 weeks after fertilization. Congenital defects of the femur range from simple hypoplasia to complete absence of bone. The clinical distinction between different types of femoral defects is important as a guide for predicting limb development.1-3 We report this case because it is a rare case and needs proper evaluation and management. The majority of cases, however, are sporadic, and therefore, the parents may be reassured that the risk of further offspring being affected is negligible. [1,2]

Case Report

A 3-month-old baby girl present with a complaint of a short stature. She was born spontaneously at 38 weeks of gestation with a birth weight of 2575 g, height 36 cm, and head circumference of 31 cm. The patient was born remarkable, 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 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 supplement and folic acid tablets. Both of the patient's parents do not smoke. There was no history of exposure to other teratogens. There were no abnormalities were found in the father's and mother's families.

The patient's condition is fit, fully breastfeed since birth. There is no fever, cough or runny nose. Patients receive routine immunizations according to schedule. Physical examination showed 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, throat found no abnormalities and no enlarged lymph nodes were found in the neck. Examination of the thorax, the heart obtained a single heart sound (S1 and S2), regular, no murmurs and gallops. In the lungs, breath sounds were vesicular, no rhonchi or wheezing were found. On abdominal examination, bowel sounds were within normal limits, there was no distension, palpable tenderness, no enlargement of the liver and spleen, and a palpable mass. Upper and lower extremities were warm, dry and red, capillary refill time was less than 2 seconds, no edema was found. The upper extremities have normal structure and function. Both of the patient's lower

extremities appear to be short in size, the patella is not palpable, both legs can be moved with a limited range of motion (Figure 1).

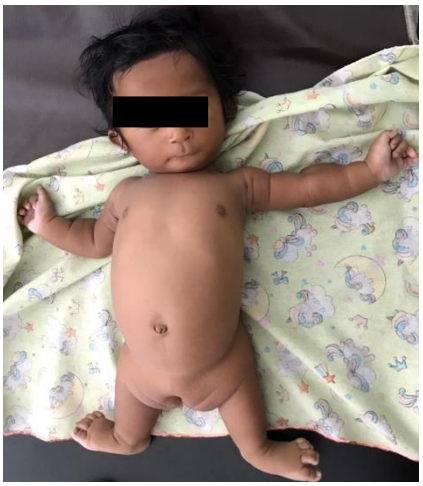

Figure 1. Clinical picture of patients

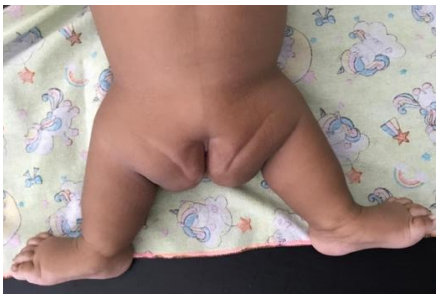

Figure 2. Bilateral femoral agenesis shown

It was further confirmed by bilateral lower extremity plain X-ray examination (Figure 3), the X-ray did not reveal bilateral femurs leading to total bilateral femur agenesis, both tibia and fibula were normal length with the proximal ends of the cruris being lateral to the bilateral acetabulum, shape and size pelvis within normal limits.

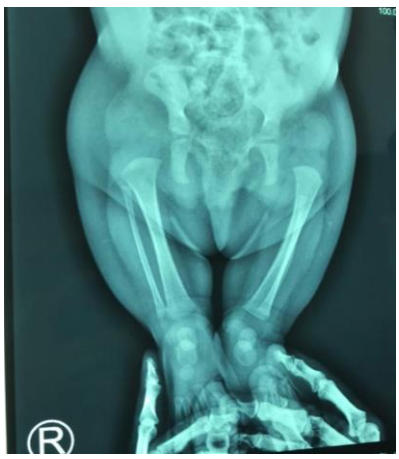

Figure 3. Femur X-ray of patient

Previously, the patient also received babygram examination at the Makassar Hospital after birth (Figure 3). On an anteroposterior chest X-ray performed immediately after the baby was born, the thorax was within normal limits, the hemivertebra culi, suspected developmental of the hip.

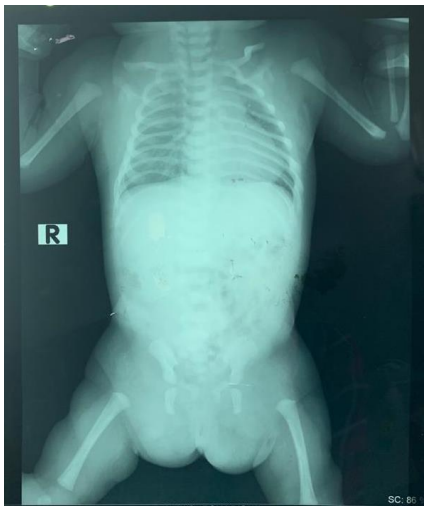

Figure 4. Babygram of patient

Discussion

Limb defects are relatively frequent congenital defects but they occur mostly in the upper extremities and are, in most of the cases minor – as polydactyly, supernumerary digits, syndactyly, fusion of two or more digits. Congenital femoral hypoplasia is a rare but complex major limb defect, ranging from simple shortening of the femur to complete femoral agenesis. Bilateral femoral agenesis is a rare and unusual anomaly and to the best of our knowledge, only six cases have so far been reported. This disorder is more common in females. These anomalies can occur singly or in associated with another anomaly such as fibular hemimelia (most common), clubfoot, an absence of lateral foot rays, congenital cardiac defects, or spinal dysplasia. Other structure anomalies are often involved; these include the acetabulum, musculature, vessels, ligaments of the knee, tibia, fibula, and foot. In our case, there are no other anomalies found. [3,4,6]

Bone ossification, or osteogenesis, is the process of bone formation. This process begins between the sixth and seventh weeks of embryonic development and continues until about twenty-five years of age; but varies for each individual. There are two types of bone ossification, intramembranous and endochondral. Each of these processes begins with a precursor of mesenchymal tissue. Intramembranous ossification directly converts mesenchymal tissue into bone and forms the flat bones of the skull, clavicle, and most of the bones of the skull. Endochondral ossification begins with the transformation of mesenchymal tissue into cartilage, which is then replaced by mature bone and forms the axial skeleton and long bones. [7,8,9]

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. Many of the genetic syndromes are associated with congenital malformations, and 0.2% of newborn have severe limb anomaly. 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. There was some of agenesis femur associated with femoral facies syndrome. Most relevant features include hypoplasia of the femurs and a characteristic facies with short nose, long philtrum, thin upper lip and micrognathia. However, none of the above mentioned factors could be identified in our case. Other frequent findings are talipes equinovarus, syndactyly and preaxial polydactyly, cleft palate and/or lip, upslating palpebral fissures and microtia. Costal, vertebral, lung, heart, renal and urinary abnormalities are also associated. [10,11,12]

The classification by Aitken is widely used. This classification is based on the severity of the hip and femur radiographic findings. There are four classes in Aitken classification: Aitken classification is descriptive but not helpful in treatment. Paley and Guardo developed the classification system for congenital femur deficiency based on the factors that influence lengthening reconstruction. The case presented here does not fulfill any classification above. [12,13]

Conclusion

Congenital femur absence is an extreme form from congenital femur deficiency. This anomaly can occur singly or in associated with another anomaly such as fibular hemimelia (most common), clubfoot, an absence of lateral foot rays, congenital cardiac defects, or spinal dysplasia. There are several classifications for this anomaly, but nothing matches to our case. Further evaluation and management of this case can help the patient and the parents to accept her condition and be able to live a good social and economically productive life.

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