Left femoral hypoplasia associated with congenital fibular agenesis – A rare case report

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ABSTRACT

Background and objectives. Unilateral or bilateral femoral hypoplasia represents a rare clinical entity and it can be isolated or associated with other limb malformations such as: congenital absence of the fibula (the most common association), the absence of lateral foot rays or clubfoot, or other organ defects such as spinal dysraphism, or congenital cardiac malformations. Only a few cases have been detected by antenatal ultrasonography so far.

Materials and methods. We present the case of a newborn admitted at birth in the Neonatology Department of County Emergency Clinical Hospital of Constanța, diagnosed prenatally with left femur hypoplasia, confirmed postnatally.

Results. The subject is a full-term female neonate, born at 38 weeks gestational age, with a birth weight of 2600 g, Apgar Score of 9. The mother is 39-year-old, Gravida-I, Para-I, with no medical history of consanguinity, genetic or congenital abnormalities, exposure to toxins, teratogen drugs, or viral infections. The 14 weeks and 16 weeks ultrasound revealed a shortened left femur, below the 5th percentile, with no other facial morphology, cardiac, neurologic, gastrointestinal, or genitourinary system abnormalities mentioned. A cesarean section was performed at 38 gestational weeks (premature rupture of membranes, breech presentation). The diagnosis was confirmed postpartum by X-ray.

Conclusions. The presented case emphasizes the importance of prenatal care and early detection of skeletal abnormalities for parental education and genetic counseling, offering an informed choice whether to continue with the pregnancy and planning for postnatal care with a multidisciplinary team: obstetrics and gynecology, neonatology, and pediatric orthopedic surgery.

Keywords: femoral hypoplasia, fibular agenesis, early detection, multidisciplinarity

INTRODUCTION

The lower extremities formation during fetal growth is the result of numerous growth factors that act in a precise sequence to induce the normal development of the limbs. During embryogenesis, between the 4th and 8th week the limb buds start to develop. This interval of intense proliferation and differentiation of structures is the period when the majority of the limb abnormalities originate, with a peak in the 5th and 6th weeks. The femur abnormalities range from hypoplasia to the complete absence of the bone. The prognosis of the subjects depends on the distinction between the clinical types of femoral abnormalities [1,2].

The association between unilateral femoral hypoplasia and ipsilateral fibular agenesis is rare, and the incidence or detection rate is not well known. The limb buds are visible through high-frequency ultrasound from the end of the seventh week of pregnancy. Biometry of the long bone is possible from the 11th week of pregnancy, and the rate of detection for the limb deficiencies in the first trimester of pregnancy is approximately 57%, according to Grande and colleagues [3,4].

Femoral abnormalities can be isolated or associated with other limb malformations such as: congenital absence of the fibula (the most common association), the absence of lateral foot rays or clubfoot, or other organ defects such as spinal dysraphism, or congeni-
tal cardiac malformations. The congenital dysgenesis of the fibula is the most common anomaly of the long bones, with an incidence of 1-2/100,000 births, often sporadic and of unknown etiology. Predominantly unilateral, it can be part of a syndrome or a multisystem entity. Achterman and Kalamchi separated fibular deficiencies in: type IA: fibular hypoplasia; type IB: absence of the proximal fibula; type II: complete fibular absence [5,6].

The incidence of congenital femoral defect is: 0.5-2/100,000 births and is associated with the absence of the fibula in 50-80% of cases. It is often unilateral and sporadic, but it can still be present in syndromes, requiring detailed analysis for fetal anomalies and genetic counseling [7,8].

The type of abnormality impacts the need and complexity of the treatment. Fibular hemimelia-affected subjects usually have mild limb length asymmetry and rarely require complex treatment. Those with significant difference in length of the limbs, usually also lack joint stability and require more intensive treatment. The therapy will have to address the limb asymmetry of varying severity, the standing problems or walk abnormalities. Mild lower limb asymmetry is usually solved by wearing a shoe insert or special shoes. Nevertheless, the majority of patients will need epiphysiodesis, leg-lengthening surgical correction, or prosthesis [9-10].

MATERIALS AND METHODS

We present the case of a neonate born to a 39-year-old patient, gravida I, para I, with no family history of genetic or congenital abnormalities, no consanguinity with husband, without a history of exposure to toxins, teratogen drugs or viral infections, non-smoker, with no significant findings at the physical examination.

RESULTS

Fourteen-week ultrasound revealed a shortened left femur, below the 5th percentile. The other measurements (biparietal diameter, head circumference and abdominal circumference) corresponded to the gestational age, and no other abnormalities were observed (Figure 1).

After the medical counseling, the mother decided to continue the pregnancy. Follow-up ultrasound examinations found that head and abdomen biometry continued to be compatible with the gestational age, left femoral hypoplasia, and left fibular aplasia. In addition, the fetus had normal facial appearance and the echography showed no malformations of the internal organs (Figure 2).

A cesarean section was performed at 38 gestational weeks (premature rupture of membranes, breech presentation, previous cesarean section). A 2600 g female newborn, with an Apgar Score of 9, was delivered. The face appeared normal; no other abnormalities were found. The diagnosis was confirmed postpartum by X-Ray (Figure 3).

DISCUSSION

Although parents prepare for their new baby by providing the most adequate postnatal care, including the best nutrition, safe environment, follow-up visits with specialists, the prenatal events have also a great impact on the baby’s wellbeing. Prenatal care should be initiated as soon as the pregnancy is diagnosed, with first trimester examinations being able
FIGURE 3. Postpartum antero-posterior view X-Ray showing femoral hypoplasia associated with fibular agenesis

to uncover most major organ deficiencies, including limb anomalies.

Diagnosing a limb abnormality by ultrasound as early as possible is essential and will also lead to a careful, detailed scan of the fetus. Unfortunately, it is sometimes difficult to diagnose a specific skeletal dysplasia only by ultrasound. Femoral digenesis appears in several conditions: Proximal focal femoral deficiency - dysgenesis of the proximal femur, Femur-fibula-ulna syndrome, Femoral hypoplasia/unusual facial syndrome (excluded, as facial profile appeared normal on the ultrasound and confirmed after birth), Limb – pelvis hypoplasia/aplasia syndrome (excluded as there were no genitalia abnormalities, no upper limb abnormalities) [9,10].

Most probable etiology of our case is femur-fibula-ulna complex (type I): various combinations of femoral or fibular defects; asymmetry; normal axial skeleton, internal organs, and facial profile; there were cases of femur-fibula-ulna syndrome described with normal development of the arms.

The prognosis mainly depends on the severity of the limb malformations and the possibility of orthopedic surgery. This syndrome is usually associated with normal intellect [11,12].

CONCLUSION

Our work underlines the importance of prompt and individualized prenatal care so that health problems of mother and fetus can be identified and addressed. Although some of these problems will not be solved prenatally, the care plan has to be initiated and whenever possible families have to be counseled and involved in decision making. Full term delivery is an important goal, as to minimize the risks for the pregnant woman and her newborn baby.

The presented case emphasizes the importance of assessing both legs and arms in the second-trimester scan, which is also an AIUM (American Institute of Ultrasound in Medicine) recommendation[4,6]. Early detection of skeletal abnormalities is important for parental education and genetic counseling. The goal is to offer an informed choice of whether to continue the pregnancy and the chance of a postnatal care plan with the help of a multidisciplinary team – obstetrics and gynecology, neonatology, and pediatric orthopedic surgery.

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