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A Case of Neonatal Birth with Congenital Bilateral Femoral Deficiency Undetected in Prenatal Ultrasound

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ABSTRACT

Background: Prenatal ultrasound plays an important role in the early and accurate evaluation of the congenital skeletal and non-skeletal abnormalities of the fetus and is effective in predicting pregnancy outcomes. Congenital femoral deficiency (CFD) is a rare complicated and non-hereditary anomaly that includes the hypoplasia of a portion of the femoral bone with shortening the lower limb.

Case report: The aim of this study was to report a rare case of neonatal CFD, which was not detected in prenatal ultrasound. In this study, we presented the case of a 28-year-old multiparous woman with the gestational age of 39 weeks hospitalized for labor pain. She had a history of two to three irregular prenatal visits and her only pregnancy ultrasound was related to late pregnancy, which reported a healthy live fetus with a cephalic presentation, which was in 35-36 weeks of gestation according to biparietal diameter (BPD) and femur length (FL). Finally, normal vaginal delivery occurred and a neonate was born with a severe bilateral CFD with deformity and immobilization of the lower limbs.

Conclusion: It seems that in order to early diagnosis and appropriate case management, it is necessary to differentiate both femoral and tibial bones in routine ultrasound more precisely plus measuring their length. The case also strongly emphasize the need for ultrasonic examination of fetal anomalies for all pregnant women.

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Introduction

Congenital Femoral Deficiency (CFD), also known as proximal femoral focal deficiency (PFFD), is a rare and non-hereditary musculoskeletal disorder that occurs once or twice per 100,000 live births (1). This disorder affects the pelvic structure and the proximal area of the femur bone and may be on one or both sides. It is associated with a change in the hip joint and a decrease in the length of the affected limb. Other defects associated with this abnormality include the displacement or instability of joint between the femur and knee, shortened tibia or fibula, and abnormalities of the lower limbs (1-3).

Although the exact etiology of the CFD is unclear; however, there are some hypotheses in this regard. The theory of sclerotome subtraction suggests that early damage to neural crest cells plays an important role in the incidence of the CFD. The histological studies regarding embryos showed the defective maturation of the chondrocytes on the growth plate. Other etiologies include the use of drugs (thalidomide), exposure of the mother to toxins during the period of organogenesis, anoxia, ischemia, radiation, infection, and mechanical pressure (4-6). The CFD has various manifestations, including a simple shortening of a seemingly normal femur to a complete absence of two-thirds of the proximal femoral, and acetabular aplasia (2, 7).

The prenatal diagnosis of this abnormality is

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still a challenge; however, its early detection can provide useful information for parents and health care providers regarding the management and therapeutic planning for this congenital deficiency. The aim of this study was to report a rare case of neonatal bilateral CFD that was not detected in prenatal ultrasound.

Case report

A 28-year-old woman with the third pregnancy and gestational age of 39 weeks was hospitalized in the maternity ward of Sina educational hospital for labor pain in 2017. The hospital is located in Karoun in Khuzestan province (south of Iran) with the birth rate of around 6,000 births per year. The case had a history of a term vaginal delivery and a stillbirth in the eighth month of gestation. In the present pregnancy, she experienced 2-3 irregular prenatal visits at the health center. The patient's only pregnancy ultrasound was related to late pregnancy period, which reported a healthy live fetus with a cephalic presentation in 35th-36th weeks of gestation according to biparietal diameter and femur length. The amount of normal amniotic fluid was reported and the placenta was considered posterior.

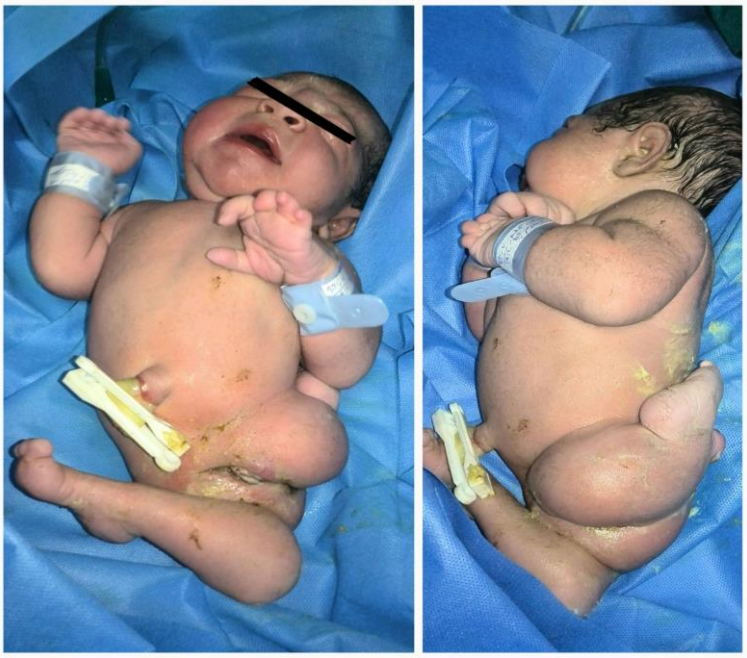

Figure1. A newborn with severe congenital femoral deficiency and lower extremity deformity

The patient had a non-consanguineous marriage and no history of medical problem, surgery, and radiography during the pregnancy. The case only used folic acid and ferrous sulfate daily. No sign of high-risk pregnancy was reported, and normal fetal movements were observed. She was hospitalized with cervical dilatation of 5cm, effacement of 50%, intact amniotic sac, along with regular and well-organized uterine contractions.

After three and a half h, normal vaginal delivery occurred and a seemingly female neonate was born suffered from severe bilateral CFD with the deformity and immobilization of lower limbs (Figure 1). The general health condition of the case was good and the first min Apgar score was 9/10. After being visited by a pediatrician, the neonate was delivered to the mother for breastfeeding.

Discussion

Musculoskeletal disorders are the second leading cause of fetal abnormalities following the defects of the central nervous system. Musculoskeletal defects have a wide range of clinical manifestations, which include the mild femoral hypoplasia, absence of functional femur, and acetabular aplasia (8, 9). This abnormality is bilateral in 15% of the patients (10). In a review of the literature, a case of CFD was reported related to a 20-year-old pregnant woman in Iran. In the evaluation of routine ultrasound, a short right femur was diagnosed for the fetus during the 26th week of pregnancy in the mentioned case study. In addition, the lengths of right femur and left femur were reported as 27 mm and 49 mm, respectively

The neonate was born with the gestational age of 38 weeks through C-section. The examination of the birth time indicated an asymmetric shortening of the lower limb and apart from that, there was no other skeletal disorders (11). The peak of the sensation of limb formation is in 5th to 6th weeks of pregnancy; therefore, major limb anomalies, such as the lack of a long bone at 7th week (12). Various etiologies, such as hereditary disorders, viral infections, radiation, focal ischemia, trauma, and chemical poisoning can be reported regarding this disorder (4, 5). Furthermore, almost half of the cases with CFD suffered from other abnormalities. A high prevalence of anomaly or complete absence of fibula bone has been reported in these individuals. The cleft lip, cleft palate, club foot, congenital heart defects, and spinal abnormalities are other abnormalities associated with CFD (11).

Recent technological advances in ultrasound imaging have been an opportunity to diagnose a large number of skeletal syndromes that need proper diagnosis for proper counseling and management. According to the literature, the prenatal diagnosis of the CFD has been noticed by means of ultrasound. This abnormality was first reported in 1989 during the prenatal period (5). Subsequently, other authors described the prenatal diagnosis of the unilateral and bilateral CFD by two-dimensional and three-dimensional ultrasounds in the weeks 18th-32th of pregnancy (13). Bronstein was able to identify and report a case with this abnormality for the first time in the 14th week of pregnancy using transvaginal ultrasound (14).

Keret described a rare case of familial CFD (15), and Hadi reported the first incidence of unilateral CFD in the fetus of an insulin-dependent diabetic woman (16). Mailath-Pokorny outlined a case of unilateral non-consanguineous CFD in the 19th week of pregnancy (17). In the present study, only ultrasound was performed in the final weeks of pregnancy, in which the abnormality was not reported. It is a fact that the size of the fetus is large at the end of pregnancy and the embryo has a limited space to move.

Moreover, the lower limbs are in a closed space; however, it is necessary to differentiate both femoral and tibia bones in ultrasound more precisely and measure their lengths (18). On the other hand, in order to diagnose early and manage these cases, it is essetial to recommend and strongly emphasize the need for ultrasound examination of the fetal anomalies for all pregnant women.

Conclusion

The CFD is a rare congenital anomaly that can be detected in precise pregnancy ultrasounds. In cases with the CFD, a targeted ultrasound should be performed for the evaluation of the face, jaw, and upper extremity. Early and accurate prenatal diagnosis provides the opportunity to manage pregnancy and plan for treatment.

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Conflicts of Interest

The authors declare no conflicts of interest.

References

1. Paley D. SUPERhip and SUPERhip2 procedures for congenital femoral deficiency. Pediatric pelvic and proximal femoral osteotomies. New York: Springer; 2018. P. 287-356.
2. Lloyd-Roberts GC. Congenital femoral deficiency. Hip Disorders in Children. London: Postgraduate Orthopaedics Series; 2013.
3. Visser JD. Limb and back reduction deficiency present at birth. Pediatric orthopedics. New York:

Springer; 2017. P. 347-392.
4. Gonçalves LF, De Luca GR, Vitorello DA, Parente LM, Philippi H, Saab Neto JA, et al. Prenatal diagnosis of bilateral proximal femoral hypoplasia. Ultrasound in Obstetrics & Gynecology. 1996; 8(2):127-130.
5. Jeanty P, Kleinman G. Proximal femoral focal deficiency. Journal of Ultrasound in Medicine. 1989; 8(11):639-642.
6. Subbarao K. Proximal femoral focal deficiency (PFFD) imaging spectrum. Journal of Research in Medical Sciences. 2015; 3(2):90-93.
7. Paley D, Guardo F. Lengthening reconstruction surgery for congenital femoral deficiency. Advanced techniques in limb reconstruction surgery. New York: Springer; 2015. P. 245-299.
8. Maldjian C, Patel TY, Klein RM, Smith RC. Efficacy of MRI in classifying proximal focal femoral deficiency. Skeletal Radiology. 2007; 36(3):215-220.
9. Cooper A, Fernandes JA. Evidence-based treatment for congenital femoral deficiency. Paediatric orthopaedics. New York: Springer; 2017. P. 71-76.
10. Apte AV, Attarde VY, Patil PV, Dahiphale D, Ahirrao M. Congenital; bilateral femoral deficiency; foetus; ultrasound, femoral length, cleft lip and cleft palate. Prenatal ultrasonographic diagnosis of bilateral proximal femoral focal deficiency with cleft lip and cleft palate. Femoral Hypoplasia/Unusual Facial Syndrome. 2016; 2(1):32-33.
11. Ghavami M, Abedinzadeh R, Ghavami S. Prenatal detection of a case of proximal femoral focal deficiency (PFFD). Iranian Journal of Radiology. 2009; 6(4):253.
12. Singh AP, Tanger R, Mathur V, Gupta AK. Unilateral isolated proximal femoral focal deficiency. Saudi Journal of Medicine and Medical Sciences. 2017; 5(2):187.
13. Otera Y, Morokuma S, Yumoto Y, Hojo S, Gotoh T, Hara T, et al. Prenatal three-dimensional images of proximal focal femoral deficiency produced by helical computed tomography. Fetal Diagnosis and Therapy. 2009; 25(3):349-353.
14. Bronstein M, Deutsch M. Early diagnosis of proximal femoral deficiency. Gynecologic and Obstetric Investigation. 1992; 34(4):246-248.
15. Keret D, Timor I. Familial congenital short femur: intrauterine detection and follow-up by ultrasound. A case report. Orthopaedic Review. 1988; 17(5):500-504.
16. Hadi HA, Wade A. Prenatal diagnosis of unilateral proximal femoral focal deficiency in diabetic pregnancy: a case report. American Journal of Perinatology. 1993; 10(4):285-287.
17. Mailath-Pokorny M, Timor-Tritsch IE, Monteagudo A, Mittal K, Konno F, Santos R. Prenatal diagnosis of unilateral proximal femoral focal deficiency at 19 weeks' gestation: case report and review of the literature. Ultrasound in Obstetrics & Gynecology. 2011; 38(5):594-597.
18. Radler C, Myers AK, Hunter RJ, Arrabal PP, Herzenberg JE. Prenatal diagnosis of congenital femoral deficiency and fibular hemimelia. Prenatal Diagnosis. 2014; 34(10):940-945.