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A Case report of Congenital Unilateral Upper and Lower Limb Reduction

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ABSTRACT

Background: Congenital limb reduction defect is a rare condition that is detectable in prenatal ultrasound. The purpose of this study was to report a rare case of congenital unilateral upper and lower limb reduction, which was not detected in prenatal ultrasound.

Case report: A primiparous woman was admitted to undergo a cesarean section due to breech presentation. The results of routine blood tests and first-trimester screening were normal. Anomaly ultrasound scan indicated the possibility of midface hypoplasia; however, three-dimensional ultrasound ruled out this condition. Other ultrasound evaluations in the following weeks of pregnancy were indicative of the normal health condition of the fetus. The outcome of the cesarean section was a girl with obvious unilateral shortening of the left limbs. Other associated anomalies included saddle nose, hepatomegaly, ichthyosis, and low-set ears.

Conclusion: Congenital limb reduction defect can be detected at an early stage through an anomaly ultrasound scan in the second trimester of pregnancy. It is recommended to allocate sufficient time for an anomaly ultrasound scan, observe all parts of the four fetal limbs, and perform fetal examination in all three coronal, longitudinal, and axial planes.

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Introduction

Congenital limb reduction defects occur when a part or the entire upper or lower limbs cannot form normally during pregnancy (1). Based on the evidence, the overall prevalence of limb reduction defects ranges from 2.5 to 7.06 per 10,000 births (2, 3). This anomaly sometimes involves only one or more limbs, which is called isolated. Sometimes, it co-occurs with one or more non-limb defects and represents a specific syndrome. However, some cases present with multiple congenital defects that are not classified in any of the known syndromes (4). In general, upper extremity defects are more common than lower extremity ones.

The upper and lower extremities are involved simultaneously in about 15% of cases. In these cases, unilateral defects are more common than bilateral defects (2). In the majority of cases, limb reduction defects are associated with other abnormalities, which include other limb defects, heart defects, and nervous and gastrointestinal disorders, respectively (5). In spite of being a rare condition, limb reduction defect is very obvious and potentially devastating to the patient and parents as it can have a significantly negative impact on patient's daily functioning and quality of life (2).

According to the guidelines, the health assessment of fetal limbs should be performed at the end of the first trimester and early second trimester of pregnancy via anomaly ultrasound scan. To confirm abnormalities diagnosed by conventional ultrasound, three-dimensional ultrasound is helpful and provides a better understanding for parents (3). Early and accurate prenatal identification of limb

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reduction defects is very helpful in pregnancy management and treatment planning. Herein, we report a case of congenital unilateral limb reduction complicated by multiple other anomalies not detected in prenatal ultrasound.

Case report

A 25-year-old nulliparous woman referred to our hospital to decide on the termination of pregnancy. She had fully received prenatal care during her pregnancy at the maternal health unit of a health center, and the results of all of her routine pregnancy tests were normal. In the first-trimester screening test, the risks of Down syndrome and trisomy 13/18 were lower than the screening cut-off. Anomaly ultrasound scan at 17 weeks and 5 days showed no health problem in the lower and upper limbs or other organs of the fetus. However, it indicated the possibility of hypoplasia of midface, thereby underscoring the need for a follow-up. The result of three-dimensional ultrasound of the fetal face that was performed 6 days later at another center was normal.

Her latest ultrasound showed a normal female fetus at the breech presentation with 39 weeks of gestation based on a biparietal diameter of 96 mm and a femur length of 77 mm. The client had no family relationship with her spouse and had not taken any medication other than ferrous sulfate, folic acid tablets, and multivitamins. At the age of 39 weeks and 2 days of gestation, the client underwent a cesarean section, which resulted in the birth of a female neonate with the first and fifth minute APGAR scores of 5/10 and 9/10, respectively. The left upper and lower limbs of the infant were smaller in appearance than the right ones, and this defect was evident in the humerus, radius, ulna, femur, tibia, fibula bones.

Figure 1. A neonate with unilateral limb reduction, ichthyosis, hepatomegaly, low-set ears, and saddle nose and the radiographic appearances

Deformity was present in the wrist and the second and third fingers of the left hand; in addition, she had left clubfoot. The palm lines on the left side of the neonate were also abnormal. However, motility was normal in the upper and lower extremities. Disseminated ichthyosis was seen in various parts of the body skin, which was more severe in some areas, such as the left foot. There were some mild erythematous areas on the skin that were greater on the left side of

the neonate's chest. Low-set and posteriorly rotated ears, saddle nose, and large mandibular bone were also observed in the neonate (Figure 1).

The biometric measurements of the case included a body weight of 3,000 g, head circumference of 35 cm, and height of 52 cm. Physical examination was clearly indicative of hepatomegaly as a result of which the lower edge of the liver was touched up to about 5 cm below the last rib. After stabilization, radiographs were taken of the neonate, which according to clinical findings, showed the shortening of all long bones of the left lower and upper extremities. Ultrasound exam showed hepatomegaly; however, other internal organs were normal.

She had no problems with urination or defecation. Echocardiography and karyotype test were requested by the physician; however, the infant's parents did not consent to perform them. The results of the neonate's blood tests were as follows: blood groop of A positive, white blood cell count of 13,200 ML, hemoglobin level of 17.2 g/dL , platelet count of 285000 /ML, blood urea nitrogen of 9 mg/dL, serum glutamic oxaloacetic transaminase of 172 IU/L, serum glutamic pyruvic transaminase of 46 IU/L, alkaline phosphatase of 302 U/L, total bilirubin of 7.6 mg/dL, direct bilirubin of 0.4 mg/dL, calcium of 10.1 mg/dL, sodium of 136 mEq/L, and potassium of 6.0 mEq/L. In addition, the test results for C-reactive protein, direct coombs, and blood culture were negative. The neonate was admitted to the neonatal intensive care unit for 4 days due to respiratory distress and discharged on the 5th day with a good general condition.

Discussion

The limb buds appear at the end of the 4th week as small bumps. The upper limb buds emerge about 2 days before the lower limbs. In the 8th week, all parts of the upper and lower limbs are completely formed. Any factors that affect the development and differentiation of the limbs at this time will cause a range of limb abnormalities. Some of these factors include genetic disorders, chromosomal abnormalities, environmental exposure, and prenatal diagnosis procedure (6). Numerous cases of limb malformations have been reported so far. In a recent study, Makvandi et al. reported a rare case of bilateral femur deficiency that was neglected in prenatal ultrasound (7). Pauleta et al. reported a case of the absence of right fibula and right tibia shortening in a 22-week-old fetus (8). In another case report in India, the absence of the left upper limb and shortness of ipsilateral femur were reported in a female infant (9).

The reported case had multiple clinical manifestations that did not fully belong to any of the known syndromes. The literature review suggests that the combination of ichthyosis and unilateral limb shortening is found in some syndromes, such as CHILD syndrome standing for congenital hemidysplasia with ichthyosiform erythroderma and limb defects. This syndrome is a very rare X-linked autosomal dominant disease characterized by unilateral limb shortening and presence of ichthyosiform erythroderma on the same side (10). In a rare case, hepatomegaly was also reported as a defect associated with this syndrome (11). There are also some cases reporting heart defects with the disease (12).

The first time that fetal limb anomalies are detectable is the first trimester of pregnancy, and the optimal time in the first trimester is after 12 weeks of gestation. The fetus should be examined in all three coronal, longitudinal, and axial planes; in addition, both left and right limbs should be examined because only one limb may be affected. Ultrasound examinations are usually abdominal and transvaginal when necessary. Evidence suggests that a combination of both methods leads to the best results (13, 14). The optimal time to detect limb reduction defects is in the second trimester of pregnancy via anomaly ultrasound scan because the uterus is completely out of the pelvis, and the fetus has grown well enough and maybe in the optimal position to be examined at this time. At the end of the second trimester and in the third trimester, due to the increase in size, it becomes more difficult to morphologically examine the fetus (3).

It is recommended that sonologists assign sufficient time to perform fetal ultrasound examination. Andrikopoulou et al. showed that an increase of less than five min in the time taken for anomaly ultrasound scan would

increase the detection rate of limb malformations more than double (15). The main benefit of the early prenatal diagnosis of limb defects is to provide parents with an opportunity to receive prenatal counseling (16). In this regard, parents are given the opportunity to discuss their child's abnormalities with various specialists. For treatable malformations, it may be necessary to collect a team of specialists for postnatal care. However, some families consider pregnancy termination for the untreatable malformations. Numerous studies have shown that the rate of pregnancy termination increases after the early diagnosis of major untreatable malformations (17, 18).

Conclusion

Unilateral limb reduction is a rare condition of congenital fetal anomalies that is detectable in ultrasound during pregnancy. The optimal time to diagnose this condition is the second trimester of pregnancy. Ultrasound examination of all parts of the four limbs of the fetus is recommended for all pregnant women. A slight increase in the amount of time spent on ultrasound improves the rate of diagnosis. An early prenatal diagnosis provides an opportunity for counseling and plays a role in deciding whether to terminate a pregnancy or continue it and seek treatment in treatable cases.

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Conflicts of Interest

The authors declare no conflicts of interest.

References

1. Vasluian E, van der Sluis CK, van Essen AJ, Bergman JE, Dijkstra PU, Reinders-Messelink HA, et al. Birth prevalence for congenital limb defects in the northern Netherlands: a 30-year population-based study. BMC Musculoskeletal Disorders. 2013; 14(1):323.
2. Kumar P, Burton BK. Congenital malformations: evidence-based evaluation and management. New York: McGraw-Hill Medical; 2008.
3. Ceausu I, Iliescu D, Poalelungi C, Posea C, Bacalbasa N, Dobritoiu D, et al. The antenatal detection of fetal limb anomalies. Congenital Anomalies: From the Embryo to the Neonate. Norderstedt, Germany: BoD–Books on Demand; 2018. P. 309.
4. Jaruratanasirikul S, Tangtrakulwanich B, Rachatawiriyakul P, Sriplung H, Limpitikul W, Dissaneevate P, et al. Prevalence of congenital limb defects: data from birth defects registries in three provinces in Southern Thailand. Congenital Anomalies. 2016; 56(5):203-208.
5. Klungsøyr K, Nordtveit TI, Kaastad TS, Solberg S, Sletten IN, Vik AK. Epidemiology of limb reduction defects as registered in the Medical Birth Registry of Norway, 1970-2016: population based study. PloS One. 2019; 14(7):1-17.
6. Gold NB, Westgate MN, Holmes LB. Anatomic and etiological classification of congenital limb deficiencies. American Journal of Medical Genetics Part A. 2011; 155(6):1225-1235.
7. Makvandi S, Kolahkaj M. A rare case of neonatal birth with congenital bilateral femoral deficiency undetected in prenatal ultrasound. Journal of Midwifery and Reproductive Health. 2019; 7(2):1712-1715.
8. Pauleta J, Melo MA, Graça LM. Prenatal diagnosis of a congenital postaxial longitudinal limb defect: a case report. Obstetrics and Gynecology International. 2010; 2010:825639.
9. Kataria U, Chhillar D. The'CHILD'Syndrome (congenital hemidysplasia with ichthyosiform erythroderma and limb defects)-a case report. Indian Journal of Research and Reports in Medical Sciences. 2014; 4(4):22-24.
10. Mi XB, Luo MX, Guo LL, Zhang TD, Qiu XW. CHILD syndrome: case report of a Chinese patient and literature review of the NAD [P] H steroid dehydrogenase-like protein gene mutation. Pediatric Dermatology. 2015; 32(6):e277-e282.
11. Yoneda K. Inherited ichthyosis: syndromic forms. The Journal of Dermatology. 2016; 43(3):252-263.
12. Bub I, Dekker P, Gericke GS. Asymmetrical limb reduction deformities--aetiological considerations. South African Medical Journal. 1984; 66(9):338-340.
13. Nicolaides KH. Screening for fetal aneuploidies at 11 to 13 weeks. Prenatal Diagnosis. 2011; 31(1):7-15.
14. Liao YM, Li SL, Luo GY, Wen HX, Ouyang SY, Chen CY, et al. Routine screening for fetal limb abnormalities in the first trimester. Prenatal Diagnosis. 2016; 36(2):117-126.
15. Andrikopoulou M, Vahanian SA, Chavez MR, Murphy J, Hanna N, Vintzileos AM. Improving the ultrasound detection of isolated fetal limb abnormalities. The Journal of Maternal-Fetal & Neonatal Medicine. 2017; 30(1):46-49.
16. Rypens F, Dubois J, Garel L, Fournet JC, Michaud JL, Grignon A. Obstetric US: watch the fetal hands.

Radiographics. 2006; 26(3):811-829.
17. Crane JP, LeFevre ML, Winborn RC, Evans JK, Ewigman BG, Bain RP, et al. A randomized trial of prenatal ultrasonographic screening: impact on the detection, management, and outcome of anomalous fetuses. American Journal of Obstetrics and Gynecology. 1994; 171(2):392-399.
18. Stoll C, Wiesel A, Queisser-Luft A, Froster U, Bianca S, Clementi M, et al. Evaluation of the prenatal diagnosis of limb reduction deficiencies. Prenatal Diagnosis. 2000; 20(10):811-818.