Bilateral Lower Extremity Agenesis: Case Report and the Importance of Transvaginal Sonography in Early Antenatal Diagnosis of Fetal Anomalies

Tuncay Nas, Pınar Özcan Keskin, Ercan Yılmaz, Aydan Biri, Fırat Mutlu, Rıfat Gürsoy

Department of Gynecology and Obstetrics, Faculty of Medicine, Gazi University, Ankara

Abstract

Background: To present bilateral lower extremity agenesis which is a rare anomaly and to point out importance of transvaginal sonography (TVS) for the antenatal diagnosis of such anomalies during early in pregnancy.

Case: A 25 year-old pregnant woman admitted to our department for first trimester nuchal translucency screening sonography. TVS revealed a 12 weeks and 2 days fetus with positive cardiac activity. Nuchal translucency was measured 1.6 mm. During sonographic fetal anatomic screening absence of bilateral lower extremities were detected. Cranium, thorax, abdominal structures and upper extremities were normal. When the family informed, termination of the pregnancy was decided. After the termination, macroscopic examination of the fetus revealed absence of lower extremities.

Conclusion: TVS is a very useful tool for the antenatal diagnosis of major fetal anomalies like extremity agenesis during the first trimester. Therefore, everyone should get benefit from this technique in early pregnancy.

Keywords: Bilateral lower extremity agenesis, antenatal diagnosis, transvaginal ultrasonography.

Bilateral lower extremity agenesis: case report and importance of transvaginal sonograpy in early antenatal diagnosis of fetal anomalies

Amaç: Nadir bir anomali olan bilateral alt ekstremite agenezi olgusunun prenatal erken tanısı ve gebelikte transvajinal sonografinin (TVS) öneminin vurgulanması.

Olgu: Yirmibeş yaşında, gravida 1, parite 0 ve antenatal takipte olan 12 hafta 2 gün gebede ense saydamlığı ölçümü için TVS de ense saydamlığı 1,6 mm tek canlı fetus izlendi TVS sırasında fetal anatomi incelendiğinde bilateral alt extremitelerin olmadığı tespit edildi. Üst ekstremiteleri, kranial yapılar, thorax, abdominal yapılar doğal olarak izlendi. Aileye gerekli bilgilendirme yapıldı ve ailenin isteği doğrultusunda gebelik sonlandırıldı. Fetusun makroskopik incelenmesinde alt ekstremitelerin total olarak gelişmediği izlendi.

Sonuç: TVS ekstremite agenezi gibi major anomalilerin antenatal tanısının mümkün olduğu 1. trimesterde anatomik yapıların görülmesi açısından son derece yararlı ve yardımcı bir yöntemdir. Bu nedenle gebeliğin bu döneminde her hekimin tereddüt etmeden rahatlıkla kullanması gerektiği kanısındayız.

Anahtar Sözcükler: Bilateral alt ekstremite agenezisi, antenatal tanı, transvajinal ultrasonografi.

Background

Ultrasonography (USG) has an importance role in obstetrics as primarily in determination of fetal anomalies. Determining major fetal anomalies within early periods of gestation is important for both family and the doctor. Mothers and fathers are curious about how is the baby during gestation, whether healthy or not and most importantly whether any structural or mental problem exists or not. The doctor also wants to know any anomaly in the beginning periods of gestation if any. Besides, the importance of the early diagnosis

Correspondence: Ercan Yılmaz, Gazi Üniversitesi Tıp Fakültesi Kadın Hastalıkları ve Doğum Anabilim Dalı
e-mail: ercanyilmazgyn@yahoo.com

increases in terms of performing it in early gestational weeks ethically and medically if the gestation will be terminated.

By the inclusion of transvaginal sonography (TVS) to obstetric practice, rate for determination of congenital anomalies in early periods of gestation increased.[1] TVS in early gestational weeks is more advantageous than transabdominal ultrasonography in determining fetal anatomy and biometry. Fetal anatomy may be scanned in detail with the help of high frequency vaginal probes; biometric measurements may be performed in early periods of gestation and structural anomalies may be diagnosed in more early periods.

USG routinely began to be used for diagnosis of structural anomalies in 18th – 20th gestational weeks in many developed countries.[2] The fetal completes the growth of organs in 12th gestational week and it can be possible to diagnose diagnosis of many malformations after that week. Furthermore, 1st trimester ultrasonographic examination may be disadvantageous when compared with 2nd trimester sonography. Being small of fetus, resolution of the device and the experience of doctor may limit optimal sonographic determination.

Due to all these reasons, TVS is not enough alone in diagnosis of fetal anomalies when performed in 1st trimester; but it increases determination of fetal anomalies when used together with 2nd trimester scanning. Bilateral lower extremity agenesis is rarely seen and its incidence 0,4/100.000.[3] Early diagnosis of these kinds of anomalies is important for deciding to continue the gestation or causing less complication if gestation will be terminated. By this study, we will present a lower extremity agenesis case diagnosed by TVS in 1st trimester.

Case

25-years-old patient having gravida 1 and parity 0 was started to antenatal process while she was pregnant for 6 weeks and 4 days as to the last menstrual period. In TVS, a single alive embryo was observed compatible with 6 weeks and 4 days. Nuchal translucency was measured as 1.6 mm. When fetal anatomy was examined during TVS, it was found that bilateral lower extremities did not exist (Figure 1). Cranium, thorax, abdominal structures and upper extremities were normal. The family was informed and the pregnancy was terminated on demand of the family. After the termination, macroscopic examination of the fetus revealed the absence of lower extremities (Figure 2).

Discussion

Bilateral lower extremity agenesis is an anomaly seen in the incidence of 0,4/100.000.3 The earliest antenatal diagnosis in the literature is reported in 12th week.[6] Ventral defects such as gastroschisis and omphalocele, gastrointestinal system anomalies such as anal atresia, pelvic hypoplasia or aplasia, urinary system anomalies, cranium

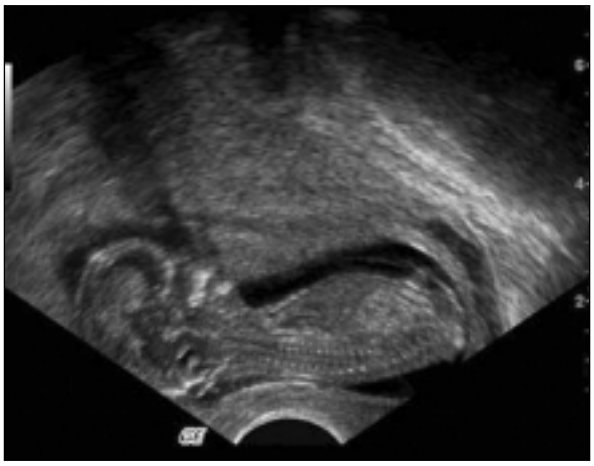

Figure 1. Ultrasonographic view of fetus.

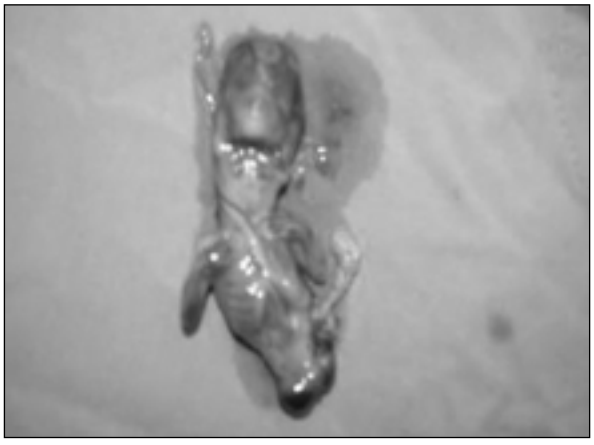

Figure 2. Macroscopic view of fetus after termination.

defects may exist along with.[5-7] Reasons such as teratogenic aspect like thalidomide, environmental factors, family background and genetic predisposition, amniotic band syndrome and vascular damage about corrion villus sampling exist in etiology. Though it is reported in some publications that similar anomalies were seen in next gestations, repetition risk in others is not reported.[12] Due to the fact that genetic predisposition may has a role in etiology, karyotype study may help diagnosis. Termination (on family's demand) in cases diagnosed easily by TVS in early period is generally correct clinical approach.

Measurement of nuchal translucency in 11th – 13th gestational weeks becomes a routine in our country as in many centers. These gestational weeks are the period that organogenesis is completed and the patient visit doctor for the measurement of nuchal translucency and the period that should be got benefit certainly for determining the important part of fatal anomalies. Even though abdominal USG is enough in that period, it is impossible for abdominal sonography to get resolution quality as TVS in many pregnants as especially in obesity patients and those who had abdominal surgery. TVS provides facility for diagnosing many anomalies such as anencephaly, megacystis, polycystic kidney, omphalocele, gastroschisis and also for choosing clinical approach in early period. Transvaginal probes with high resolution provide monitoring of early fetal growth.[13-14] Diagnosing to fetal structural anomalies by FTS is reported as 41-65% in the literature.[12-14] Therefore, TVS anomaly scanning in early period becomes a routine. In this case, the importance of TVS appeared as a possibility to observe in detail in which phase the lower extremity is insufficient and as eliminating anomalies that may occur.

Although many fetal anomalies can be diagnosed by TVS in early period, the position of 2nd trimester anomaly scanning did not change. For instance, many anomalies such as hydrocephaly, duodenal atresia appear after 2nd trimester.[13] Sonographies performed in both periods are not alternative approaches for each other. But some organs like extremities may be observed in early periods as 11th gestational week.[11] If it is possible to establish these anomalies in early period, to monitor a gestation which can be found in early period anomaly until 2nd trimester and to wait establishing the anomaly in that period is not a situation to be preferred by both family and the doctor.

As a result, anomaly scanning in 1st trimester is better and easier by TVS. Thus, we think that it is a method should be used by every doctor rather frequently and easily.

References

1. Timor-Tritsch, I.E., Blumenfeld, Z., Rottem, S. Sonoembryology. In: Timor-Tritsch, I.E., Rottem, S.(Eds). Transvaginal Sonography, New York: Elsevier. 1991, p:225-98.
2. Pilu G, Nicolaides KH (1999). Standard views for examination of the fetus. In Diagnosis of fetal abnormalities: The 18–23 Weeks Scan, Pilu G, Nicolaides KH (eds). Parthenon Publishing: London. 1999, p:3-4.
3. Rishsingani A, Yankowitz J, Mazursky J, Williamson R. Prenatal ultrasound diagnosis of amelia. Prenat Diagn 1995; 15(7): 655-9.
4. Hernadi L, Torocsick M. Screening for fetal anomalies in the 12th week of pregnancy by transvaginal sonography in an unselected population. *Prenat Diagn* 1997; 17: 753-6.
5. Ghosh G, Gupta S. Amelia with anorectal and external genital atresia. *Indian Pediatr* 2004; 41(12):1267.
6. Olney RS, Hoyme HE, Roche F, Ferguson K, Hintz S, Madan A. Limb/pelvis hypoplasia/aplasia with skull defect (Schinzel phocomelia): distinctive features and prenatal detection. *Am J Med Genet* 2001; 103(4): 295-301.
7. Cserni G, Tanko A. Limb-body wall malformation complex: an unusual developmental abnormality of the abdominal wall. Case report, clinicopathological and etiological implications. *Orv Hetil* 1997; 138(15): 931-7.
8. Froster-Iskenius UG, Baird PA. Amelia: incidence and associated defects in a large population. *Teratology* 1990; 41(1): 23-31.
9. Timor-Tritsch IE, Peisner DB, Raju S. Sonoembryology: An organoriented approach using a high-frequency vaginal probe. *J Clin Ultrasound* 1990; 18: 286-98.
10. Rosati P, Guariglia L. Transvaginal fetal biometry in early pregnancy. *Early Hum Dev* 1997; 49: 91-6.
11. Economides DL, Braithwaite JM. First trimester ultrasonographicdiagnosis of fetal abnormalities in a low-risk population. *Br J Obstet Gynaecol* 1998; 105: 53-7.
12. Whitlow BJ, Chatzipapas IK, Lazanakis ML, Kadir RA, Economide DL. The value of sonography in early pregnancy for the detection of fetal abnormalities in an unselected population. *Br J Obstet Gynaecol* 1999; 106: 929-36.
13. Rottem S, Bronshtein M. Transvaginal sonographic diagnosis ofcongenital anomalies between 9 weeks and 16 weeks, menstrual age. *J Clin Ultrasound* 1990; 18: 307-14.
14. Gaffney G, Manning N, Boyd PA, Rai V, Gould S, Chamberlain P. Prenatal sonographic diagnosis of skeletal dysplasias: A report of the diagnostic and prognostic accuracy in 35 cases. *Prenat Diagn* 1998; 18: 357-62.