Limb Body Wall Complex: First Trimester Ultrasonographic Diagnosis of a Case with Exencephaly and Megacystis

Mert TURĞAL[1], Aslıhan YAZICIOĞLU[1], Özgür ÖZYÜNCÜ[1], M. Sinan BEKSAÇ[1]

Ankara, Turkey

ABSTRACT

Limb Body Wall Complex (LBWC) is a rare developmental anomaly which is classified together with midline defects. Despite the anomaly arises from an error occuring during the embryonic folding, the exact pathophysiological mechanisms are poorly understood. The clinical picture is characterized by thoraco-abdominoschisis, eventration of the internal organs, limb reduction defects and sometimes craniofacial abnormalities. In this report we present (with the informed consent of the patient) the first trimester ultrasonographic diagnosis of a fetus with the classical findings of LBWC accompanied by exencephaly, megacystis and kyphoscoliosis which are rare in the literature. Today, this mortal anomaly can be diagnosed during the early weeks of gestation by ultrasonography. Prenatal detection of the anomaly is critical since offers an option of pregnancy termination.

Keywords: Limb body wall complex, Ultrasonography, Exencephaly, Megacystis

Gynecol Obstet Reprod Med 2015;21:106-108

Introduction

Limb Body Wall Complex (LBWC) is a rare developmental anomaly. The disorder is classified together with midline defects such as omphalocele, gastroschisis, and Cantrell Pentalogy. LBWC is characterized by thoraco-abdominoschisis, eventration of the internal organs, limb reduction defects and sometimes craniofacial abnormalities.[1] It has an incidence of 1/14,000-1/42,000 among pregnancies.[2,3] Herein, we present the first trimester ultrasonographic diagnosis of a classical case of LBWC accompanied by rarely occuring exencephaly, megacystis and kyphoscoliosis.

Case Report

A 21-year-old, primigravid woman admitted to our department with a complaint of suspected fetal anomaly at her 14th weeks of gestation. Transabdominal ultrasound revealed a fetus having several anomalies with a crown-rump length (CRL) of 80 mm which was compatible with 13 weeks 6 days. Subsequently, a transvaginal ultrasonography is performed. Transvaginal ultrasonographic examination revealed ectopia cordis and a large midline thoracoabdominal defects containing liver-bowel herniation, exencephaly, megacystis and kyphoscoliosis (Figure 1-2). The diagnosis of LBWC or a variant of Cantrell Pentalogy was considered according to the current clinical picture. Parents were informed about the poor prognosis; after obtaining the informed consent, termination of pregnancy was performed with vaginally administered misoprostol. Macroscopic evaluation of the fetus was compatible with his antenatal ultrasonography. Chorionic villus sampling (CVS) result was normal, 46, XY. Autopsy revealed thoraco-abdominoschisis, externally located heart, liver and intestines, exencephaly, megacystis, thickness of the left lower extremity possibly due to an amniotic band and foot amputation, fusion of the fingers of both hands and right foot and flexion contractures of bilateral wrists (Figure 3).

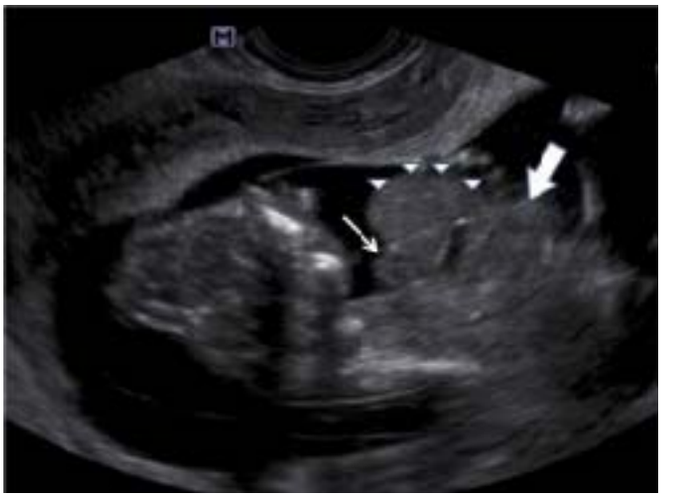

Figure 1: Sagittal ultrasonographic view showing fetal heart (white thin arrow), liver (arrow heads), and intestinal structures (white thick arrow).

[1] *Department of Obstetrics and Gynecology Hacettepe University Medical Faculty, Ankara*

Address of Correspondence: *Aslıhan Yazıcıoğlu*
Department of Obstetrics and Gynecology Hacettepe University Medical Faculty Ankara, Turkey
draslihanakar@hotmail.com

Submitted for Publication: *04. 02. 2014*
Accepted for Publication: *14. 03. 2014*

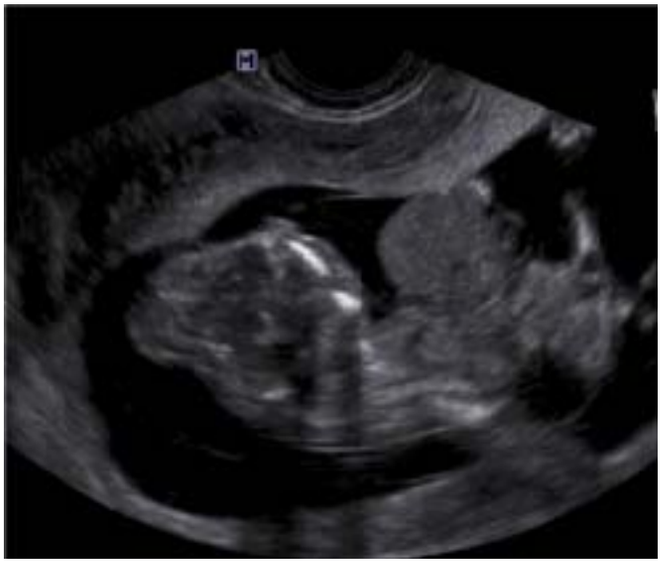

Figure 2: Megacystis appearence of the fetus with LBWC

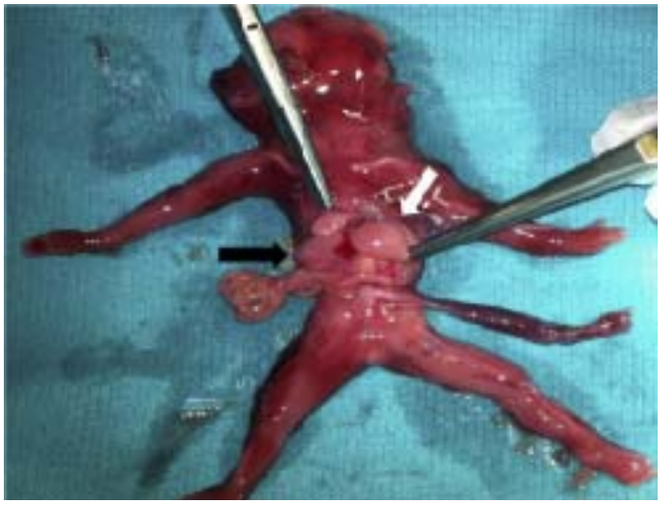

Figure 3: Macroscopic examination of the fetus after termination of pregnancy. Please note the presence of large thoracoabdominal defect, syndactyly, amputated fingers, and exencephaly (black arrow; liver, white arrow; heart)

Discussion

LBWC is a developmental abnormality which occurs during three-directional by means of cephalic, caudal and lateral directions, embrionic folding phase of embrionic development.[4] This anomaly is characterized by thoraco-abdominoschisis and eventration internal organs.[1] Furthermore, these fetuses may present with kyphoscoliosis, short umbilical cord, limb reduction defects, craniofacial abnormalities.[5] Although environmental and genetic factors are thought to play a role in the pathophysiology, it has not been clarified yet. Defects of embrionic folding, early ruptures of amniotic membrane at 4-6th weeks of gestation and secondary defective embrionic development are thought to be responsible.[6] In addition, deficient formation of ectodermal structures during early embrionic disc phase is also blamed.[7]

The diagnosis of LBWC is often made during second trimester, however cases detected during the first trimester screening are increasing in frequency.[6-8] The abnormality is encountered one in 7, 500 pregnancies in the first trimester, and 71.4% of these fetuses demonstrate increased nuchal translucency (NT), above 95. percentile.[9]

In the medical literature, karyotype analysis were reported to be normal. Furthermore, 2 cases were reported to have abnormal karyotype results. These abnormalities were uniparental disomy maternal in origin and mosaic trisomy 2 possibly originating from plasenta, both were diagnosed by CVS.[10,11] We performed CVS and karyotype was reported to be normal, consistent with the current literaure.

Fetuses with this developmental defect may have accompanying anomalies. Ninety-five percent of cases have limb defects and 75% have kyphoscoliosis or lordosis.[12] In addition, midline defects such as short umbilical cord, exencephaly and encephalocele may be encountered. The case presented here demonstrated frequently accompanying abnormalities of limbs and kyphoscoliosis with exencephaly and megacystis. Although there are several case reports of exencephaly in the literature, there is only one report of megacystis.[13]

In conclusion, LBWC is a very rare anterior abdominal wall defect with poor prognosis. As presented in this case, basic fetal anatomic evaluation in the first trimester is important in the early diagnosis of fetal anomalies which are incompatible with life.

Ekstremite Vücut Duvarı Kompleksi: Ekzensefali ve Megasistis Bulunan Olgunun İlk Trimester Ultrasonografik Tanısı

ÖZET

Ekstremite vücut duvarı kompleksi nadir görülen bir gelişimsel anormallik olup orta hat defektleri ile birlikte sınıflandırılmaktadır. Bu anomali, embriyonik katlanma sırasında meydana gelen bir hata sonucu ortaya çıkmakla birlikte patofizyolojik mekanizma tam olarak aydınlatılamamıştır. Tablo torako-abdominoşizis, iç organların eventrasyonu, ekstremite redüksiyon anomalileri ve bazen kranyofasyal defektler ile karakterizedir. Burada, (hastanın bilgilendirilmiş onamı ile birlikte) klasik ekstremite vücut duvarı kompleksi bulgularına ek olarak literatürde nadir olan ekzensefali, megasistis ve kifoskolyozu bulunan bir fetusun ilk trimesterdeki ultrasonografik tanısı sunuldu. Günümüzde, yaşamla bağdaşmayan bu anomalinin erken gebelik haftalarında ultrasonografi ile tanısı mümkündür. Anomalinin prenatal dönemde tespit edilmesi aileye gebeliğin sonlandırılması seçeneğini sunması nedeniyle önem taşımaktadır.

Anahtar Kelimeler: Ekstremite vücut duvarı kompleksi, Ultrasonografi, Ekzensefali, Megasistis

References

1. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. Am J Med Genet 1987;28(3): 529-48.
2. Kurosawa K, Imaizumi K, Masuno M, Kuroki Y. Epidemiology of limb-body wall complex in Japan. Am J Med Genet 1994;51(2):143-6.
3. Chen CP, Lin CJ, Chang TY, Hsu CY, Tzen CY, Wang W. Second-trimester diagnosis of limb-body wall complex with literature review of pathogenesis. Genet Couns 2007;18(1):105-12.
4. Lockwood CJ, Scioscia AL, Hobbins JC. Congenital absence of the umbilical cord resulting from maldevelopment of embryonic body folding. Am J Obstet Gynecol 1986;155(5):1049-51.
5. Costa ML, Couto E, Furlan E, Zaccaria R, Andrade K, Barini R, et al. Body stalk anomaly: adverse maternal outcomes in a series of 21 cases. Prenat Diagn 2012; 32(3):264-7.
6. Sepulveda W, Wong AE, Simonetti L, Gomez E, Dezerega V, Gutierrez J. Ectopia cordis in a first-trimester sonographic screening program for aneuploidy. J Ultrasound Med 2013;32(5):865-71.
7. Murphy A, Platt LD. First-trimester diagnosis of body stalk anomaly using 2- and 3-dimensional sonography. J Ultrasound Med 2011;30(12):1739-43.
8. Tsirka A, Korkontzelos I, Diamantopoulos P, Tsirkas P, Stefos T. Prenatal diagnosis of body stalk anomaly in the first trimester of pregnancy. J Matern Fetal Neonatal Med 2007;20(2):183-4.
9. Daskalakis G SN, Jurkovic D, Snijders RJ, Nicolaides KH. Body stalk anomaly at 10-14 weeks of gestation. Ultrasound Obstet Gynecol 1997;(10):416-8.
10. Chan Y, Silverman N, Jackson L, Wapner R, Wallerstein R. Maternal uniparental disomy of chromosome 16 and body stalk anomaly. Am J Med Genet 2000;94(4):284-6.
11. Smrcek JM, Germer U, Krokowski M, Berg C, Krapp M, Geipel A, et al. Prenatal ultrasound diagnosis and management of body stalk anomaly: analysis of nine singleton and two multiple pregnancies. Ultrasound Obstet Gynecol 2003;21(4):322-8.
12. Vural M, Bayar Ü, Başaran M, Barut A, Ungan B, Akbulut V. Prenatally Diagnosed Limb Agenesis in A Case of "Limb Body Wall Complex" At 18th Week of Gestation. Turkiye Klinikleri J Gynecol Obst 2008;18(1):72-4.
13. Chen CP, Hsu CY, Wu PC, Tsai FJ, Wang W. Prenatal ultrasound demonstration of limb-body wall complex with megacystis. Taiwan J Obstet Gynecol 2011;50(2):258-60.