

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

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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 occurring 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

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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 occurring 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 contain-

ing 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).

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Figure 2: Megacystis appearance 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, embryonic folding phase of embryonic 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 embryonic folding, early ruptures of amniotic membrane at 4-6th weeks of gestation and secondary defective embryonic development are thought to be responsible.⁶ In addition, deficient formation of ectodermal structures during early embryonic 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 placenta, both were diagnosed by CVS.^{10,11} We performed CVS and karyotype was reported to be normal, consistent with the current literature.

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.

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

ÖZET

Ekstremitte 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, ekstremitte redüksiyon anomalileri ve bazen kranyofasial defektler ile karakterizedir. Burada, (hastanın bilgilendirilmiş onamı ile birlikte) klasik ekstremitte 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: Ekstremitte vücut duvarı kompleksi, Ultrasonografi, Ekzensefali, Megasistis

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