Amniotic Band Syndrome and a Rarely Seen Subtype: Limb Body Wall Complex

Amniyotik Bant Sendromu ve Nadir Görülen Bir Subtipi: Ekstremite Vücut Duvar Kompleksi

Amniyotik Bant Sendromu Subtipi: Vücut Ekstremite Duvar Defekti / Amniotic Band Syndrome Subtype: Limb Body Wall Complex

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Özet

Amniyotik Bant Sendromu, fibröz amniyotik bantların fetal dokuları sıkıştırması ile ortaya çıktığı düşünülen çok çeşitli fetal konjenital anomalileri kapsayan nadir bir konjenital bozukluktur. Hastalardaki semptomlar minör parmak ampütasyonlarından fatal konjenital anomalilere kadar uzanan geniş bir spektrumda izlenir. Erken ikinci trimesterde prenatal ultrasonografide belirli ultrasonografik bulguları ile amniyotik bant sendromunun ciddiyeti saptanabilir. Amniyotik bant sendromunun fatal bir subtipi olan ekstremite vücut duvar kompleksinin erken dönemde tanınması hastaların bilgilendirilmesi ve erken dönemde terminasyonuna olanak sağlar. Biz burada erken ikinci trimesterde tanısını koyduğumuz ve termine ettiğimiz vücut ekstremite duvar kompleksine sahip bir olguyu sunmak ve prenatal ultrasonografik bulgularını tartışmayı amaçladık.

Anahtar Kelimeler

Amniyotik Bant Sendromu; Fetal Anomali; Prenatal Ultrasonografi

Abstract

Amniotic band syndrome is a rare congenital disorder that is caused by intrauterine entrapment of fetal parts in fibrous amniotic bands. Patients may have varying clinical features ranging from minor digital amputations to fatal congenital anomalies. Prenatal ultrasound scan regarding the specific findings of amniotic bant sendromu during the early second trimester may provide the detection of the severity of the disease. Because Limb Body Wall Complex is a lethal condition, earlier diagnosis may also provide to counsel the parents and terminate the pregnancy in an earlier period. We herein present a case of Limb Body Wall Complex, diagnosed and terminated at 14 weeks of gestation and discuss the ultrasonographic findings.

Keywords

Amniotic Band Syndrome; Fetal Anomaly; Prenatal Ultrasonography

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Introduction

Amniotic band syndrome (ABS) is a rare congenital disorder, thought to develop secondary to the compression of fetal tissues by fibrous amniotic bands. Its incidence is one out of 1,200 to 15,000 live births [1]. It is a sporadic condition and does not have genetic origin. Two main pathogenesis has been introduced to explain the pathogenesis of this disease; one is the exogenous theory that proposed the early rupture of the amniotic membrane leading to fibrous bands. These fibrous bands usually entrap a part of extremity, reduce the blood circulation and lead to autoamputation of a digit or entire limb in utero [2]. The other endogenous theory proposes the vascular problems as a cause of the condition [3]. Although the most common finding of this syndrome is digital amputation, it may not be seen in all cases [4]. Other observed anomalies are as follows; contraction rings, lymphedema, craniofacial defects, encephalocele, anencephaly, drop foot and multiple joint contractures [5]. Although the diagnosis is possible in ABS by prenatal ultrasonography, the fetuses are usually diagnosed by postpartum physical examination [6]. We aimed to present a case of a Limb Body Wall Complex (LBWC), the fatal subtype of ABS, which was diagnosed during the early second trimester (14th weeks of gestation) ultrasound scan.

Case Report

A 31-year-old G1 case was referred to our early pregnancy clinic with the suspicion of multiple fetal abnormalities. Her medical history revealed no prior systemic diseases and drug use, no history of infection or trauma during pregnancy, and no consanguineous marriage. The ultrasonography examination revealed amniotic fluid in normal ranges, and the presence of a live fetus with a CRL of 80 mm, compatible with 14 weeks. Fetal movements were free at the lower extremities, but the upper extremity and the head were noted not to move extensively. The detailed ultrasonography examination revealed wrapping amniotic bands, especially the left hand and around the head of the fetus, meandering as loose fibers in the amniotic cavity. The upper right extremity of the fetus was seen to be totally absent starting from the shoulder level. Both lower extremities on the other hand were observed to be in normal dimensions and moving freely. The fetal cranium was seen to be totally absent above the bilateral orbita at the anterior and occipital bone at the posterior site and the brain tissue was seen to be free in the amniotic fluid (Figure 1). An abdominal wall defect at the umbilicus in the fetal anterior wall and fetal intestines expulsed out to the amniotic cavity through this defect were observed (Figure 2). The diagnosis with those findings was 'LBWC' and

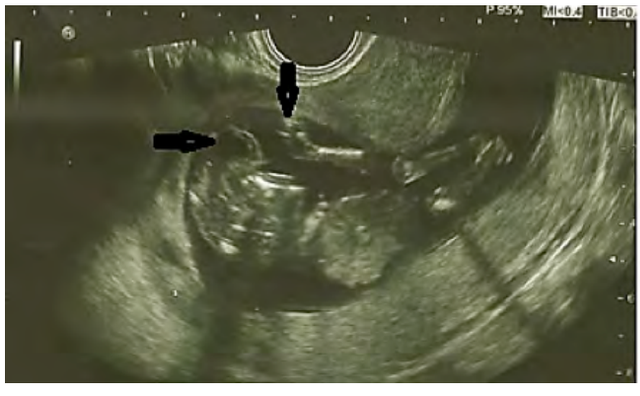


Figure 1. Ultrasound scan performed at 14 gestational weeks. Left arrow points at the fetal exencephaly. Right arrow points the left hand in a fixed extended position due to entrapment of the amniotic bands.

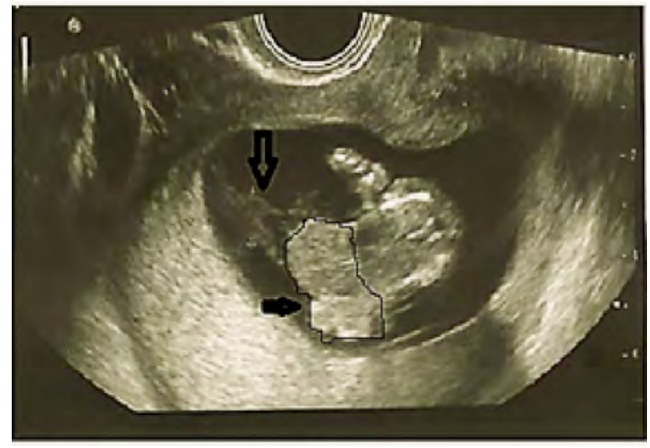


Figure 2. Ultrasound scan performed at 14 gestational weeks. Thin arrow points the protrusion of the fetal abdominal organs through the thoraco-abdominal wall defect. Thick arrow points the attachment between these structures and fetal amniotic membranes.

detailed information was given to the patient about the condition of the fetus and possible perinatal results of the condition. Upon the request by the patient to terminate the pregnancy, she signed for approval and a medical termination protocol was applied. Upon morphological examination of the fetus after the abortus, distal phalangeal amputations were observed in the left hand on the fourth and fifth fingers, while the right upper extremity was totally absent (Figure 3). A cranial defect starting



Figure 3. A photograph of fetus taken after abortion. Thin arrow points the amniotic band around the fetal left hand and absence of fetal cranium above the level of orbita. Thick arrow points the wide asymmetrical thoraco-abdominal wall defect.

from the bilateral fetal orbita to the occipital bone and a disoriented cortex not covered with calvarium and an anterior wall defect starting from the anterior fetal abdominal wall extending to the left axilla including the thoracic wall. The intestines were observed to be expulsed through this defect and they were seen outside the abdomen (Figure 4).

Discussion

The case reported herein is compatible with the diagnosis of LBWC, which is a fatal subtype of amniotic band syndrome. The diagnosis of LBWC requires at least two of the following three criteria, which are exencephaly and facial clefts, thoracic and/or abdominoschisis, and extremity defects [7]. The case presented herein meets all three criteria. In addition to the upper extrem-

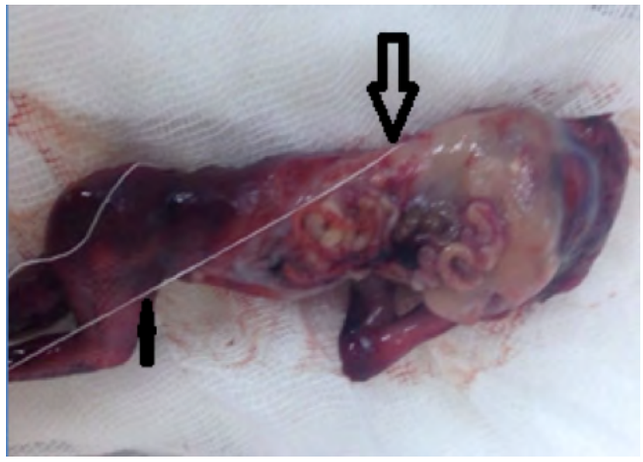


Figure 4. A photograph of fetus taken after abortion. Thick arrow points the absence of left upper extremity and expulsion of fetal intestines through the fetal body wall defect. Thin arrow points the white fetal amniotic bands surrounding the fetal body.

ity abnormalities in this case, both a craniofacial defect and an atypical wall defect including both the thorax and abdomen were present. Internal organ defects are generally present in 95% of the cases of LBWC and more than 70% of those defects are thought to develop secondary to vascular disorders [8]. However, since the case presented herein was diagnosed at an earlier stage of pregnancy and since no postmortem pathological study was performed, information about the internal organ defects is not provided.

Other non-lethal conditions that might be confused with LBWC are polymalformation complexes such as Cantrell Pentalogy, omphalocele-exstrophy-imperforated anus-spinal defects, and isolated gastroschisis [9]. Five typical findings necessary to be present for the diagnosis of Cantrell Pentalogy are cardiac abnormalities such as ectopic cordis, lower sternal defects, midline supraumbilical wall defects, anterior diaphragmatic defects, and defects of the diaphragmatic segment of the pericardium. On the other hand, ultrasonography findings of omphalocele–exostrophy–imperforated anus-spinal defects complex are absence of fetal bladder, presence of an infraumbilical abdominal wall defect, spinal distortion, and a sacral myelomeningocele [10]. In contrast to LBWC, the fetal intestines are covered with peritoneum in the two syndromes stated above. The fetal intestines are freely present in the amniotic cavity in gastroschisis; however, there is no additional wall defect.

Since LBWC is a lethal condition, early diagnosis of this syndrome provides an opportunity to terminate the pregnancy in an earlier period. Therefore it should be differentiated from other syndromes with a careful prenatal ultrasonography as early as possible. Therefore, obstetricians should carefully examine the fetus regarding these specific ultrasonographic findings during the early second trimester in order to counsel the parents with an early diagnosis of LBWC.

Competing interests

The authors declare that they have no competing interests.

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