

Prenatally Diagnosed Limb Body Wall Complex

Yakup KUMTEPE, Bünyamin BÖREKÇİ, Metin İNGEÇ, Sedat KADANALI

Department of Obstetrics and Gynecology, Faculty of Medicine, Atatürk University, Erzurum, Turkey

Abstract

In this report, a case of limb body wall complex (LBWC) diagnosed by ultrasonography is presented. Differentiation of limb body wall complex from abdominal wall defects and evaluation of limb body wall complex in view of literature is cited. Ultrasonographic detection of abdominochisis, scoliosis, and abnormalities of the lower extremities is important for prenatal diagnosis of LBWC.

Key words: limb body wall complex, abdominal wall defects, amniotic bands

Özet

Limb Body Wall Complex Defektlerinin Prenatal Ultrasonografik Tanısı

Bu çalışmada ultrasonografik olarak tanısı konan bir limb body wall complex (LBWC) vakası sunulmaktadır. Çalışmada limb body wall complex ile abdominal duvar defektleri ayırımının yapılması ve literatür ışığında LBWC'nin değerlendirilmesi amaçlanmıştır. Amdominochisis, skolyoz ve alt ekstremitte anormalliklerinin sonografik olarak saptanması LBWC'nin prenatal tanısında önemli bir yer tutmaktadır.

Anahtar sözcükler: limb body wall complex, karın duvarı defektleri, amniotik bant

Introduction

Limb body wall complex has been consisted of the spectrum of defects, seen early amnion rupture sequence. It has been proposed that this disorder occurs from early amnion rupture with direct mechanical pressure and amniotic strands producing the typical defects (1,2,3). Besides, it was reported that this complex occurs as a result of defect in germ disc causing abnormal formation in amniotic cavity, or this complex is seen as a result of teratogenic effect during early pregnancy (4). It was also pointed out that this complex occurs as a result of vascular abnormality during early embrional period (4). Van Allen et al.(4), described this defect as existence of three criteria; exencephaly or encephalocele with facial clefts; thoraco and/ or abdominoschisis; and limb defects. The diagnosis was based on the presence of two out of three above abnormalities. On the other hand, Pagon et al., described that limb body wall complex is a fetal malformation and consists of neural tube defects, body wall disruption and limb abnormalities (5). The diagnosis is made by the presence of at least two of the above features (6). The prognosis of LBWC is very poor when compared with the isolated omphalocele or gastroschisis. The pathogenesis of malformations is still unknown, although various causative theories, including amniotic band (3),

vascular disruption (4,5) and embryonic dysplasia, (6) have been proposed. (1) Martinez- Frias, determined the presence of body wall defects with evisceration of thoracic and/or abdominal organs and other congenital abnormalities with or without limb deficiencies is considered to be the LBWC (8).

Case Report

A.D, 22-year-old, applied to routine pregnancy information to our hospital. 18-weeks pregnancy and multiple anomalies were determined in ultrasound examination. Since the presence of gastroschisis and extremity abnormalities in ultrasonography, it is thought that it might be LBWC. For determination of probable chromosomal aberrations amniocentesis was applied. After amniocentesis, the patient was subjected to cytogenetic diagnosis. Karyotype was reported as 46, XX and medical abortion was made with decision of parents. In post mortem physical examination, fetus showed same growth with gestational age, and extremity anomalies, gastrochisis have called attention, especially craniofacia was found to be normal. There were defects on abdominal right antero-lateral wall and it was clearly seen intestinal ans and liver from these defects (Figure 1). Bottom of lumbosacral part of vertebral colon and low extremities were deviated to left side when the body was checked from the backside (Figure 2). The relationship between femur and hip were found to be abnormal and it was not in normal anatomic localisation. In right, low extremity webbing in knee, metatarsus adductus, deformities of equin and four fingers were determined. On the other hand, flexion deformity and pas equinovarus were found in low left extremity, but there was no

Corresponding Author: Yakup Kumtepe, MD
Terminal Cad. No: 14/2, Amil Ap. A Blok,
25200, Erzurum, Türkiye
Phone: +90 (442) 218 66 06, +90 (533) 652 66 14
Fax: +90 (442) 316 63 33
E-mail: ykumtepe@hotmail.com



Figure 1. A view of abdominal wall defects (intestinal ans and liver) from anterior abdominal wall.

webbing in knee unlike the other extremity. There was a fibrous tissue beginning from wrist and ending at placenta, and its diameter was 3-4 mm (Amniotic band). Genital structure was described as a normal female.

Discussion

In our case, there were anomalies in extremities and abdominal wall defects, especially gastroschisis have called attention. LBWC was diagnosed as a result of abdominal wall and limb defects. Scoliosis was determined at the lumbosacral part of vertebral colon. Van allen et al., in their study, described this defect as existence of three criteria; exencephaly or encephalocele with facial clefts; thoraco and/ or abdominoschisis; and limb defects (4). They also pointed out that this complex took place as a result of vascular abnormality during early embrional period. Our case, having abdominal wall and limb defects, conformed to Van Allen et al., proposes that the diagnosis of LBWC should be based on the existence of two out of three of anomalies determined (4). Negishi et al. prenatally diagnosed eight fetuses by ultrasonography as having LBWC on the basis of several combined abnormalities, examined six of the eight fetus for chromosomal constitution, and found that all of them were karyotypically normal (6). In our case, it was also found that all karyotype was reported to be normal, 46xx as reported by Van Allen et al. (4) and Negishi et al. (6).

For the differential diagnosis between body wall defects of LBWC and abdominoschisis, early diagnosis of limb body wall complex play important role since prognosis is very poor, unlike that of other abdominal wall defect, and termination must be applied. This situation is important for early termination of pregnancy. No familial case has been reported related to limb body wall complex and the pathogenesis of malformations is



Figure 2. A view of vertebral coloumn defects and lower extremities from backside.

still unknown. Viscarello et al. (7) reported that two cases of LBWC were related to cocaine abuse and Negishi et al. (6) diagnosed this complex in one of their eight cases examined. Among the different proposed theories concerning the etiology, the following three seem to be major ones: amniotic band, (1-3) vascular disruption, (3-5) and embryonic dysplasia (3) In our case, there was an amniotic band with 3-4 mm, so our case is related to amniotic band etiology.

Conclusion

Ultrasonographic detection of amdominoschisis, scoliosis, and abnormalities of the lower extremities is important for prenatal diagnosis of LBWC. It is possible that LBWC might be confused with abdominal wall defects. Since, while limb body wall complex has a very poor prognosis, abdominoschisis could be recovered by operation.

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