CASE REPORT

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Prenatal Sonographic Diagnosis of Limb-Body Wall Complex: One Case of a Rare Congenital Anomaly

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ABSTRACT Limb Body Wall Complex is also known as the body-stalk syndrome. It is a rarely seen clinical entity. In this case report we represent a 30 year old woman with 18 weeks of pregnancy associated with rare congenital anomaly "limb-body wall complex" also known as "body stalk syndrome" with prenatal ultrasonographic diagnostic features and immediate after delivery evaluation.

Keywords: Limb-body wall complex; omphalocele; gastroschisis; body stalk syndrome; single umbilical artery; ectopia cordis; scoliosis; multiple amniotic band; club foot

Limb-body wall complex (LBWC) is a rare, complicated, polymalformative fetal syndrome with essential features of:

Exencephaly and/or encephalocele with facial clefts,

Thoraco and/or abdominoschisis, and

Limb defects.

The sonographic hallmarks of LBWC are neural-tube abnormalities, severe scoliosis, positional deformities, and abnormalities of fetal membranes.[1] Generally, the diagnosis is based on any 2 of the 3 previously mentioned features. Two adhesion phenotypes have been described, the "placentocranial" and "placentoabdominal." LBWC is also known as "body stalk syndrome." There is no treatment for LBWC. Lethal condition either in utero in early neonatal period. Body stalk anomaly is an accepted fatal anomaly.[2] Therefore its early diagnosis helps in proper management of the patient. LBWC should be diagnosed antenatally and terminated.

CASE REPORT

A 30 year old G2P1 female was referred to our hospital. The ultrasonography revealed male fetus with gestational age of 18 weeks using biparietal diameter (BPD). However it revealed a large abdominal wall defect with intestinal coils and liver seen herniating into the amniotic cavity. Spinal dysraphism was also seen in this fetus. Multiple amniotic bands were present (Figure 1). Ectopia cordis was detected. Color Doppler study was performed

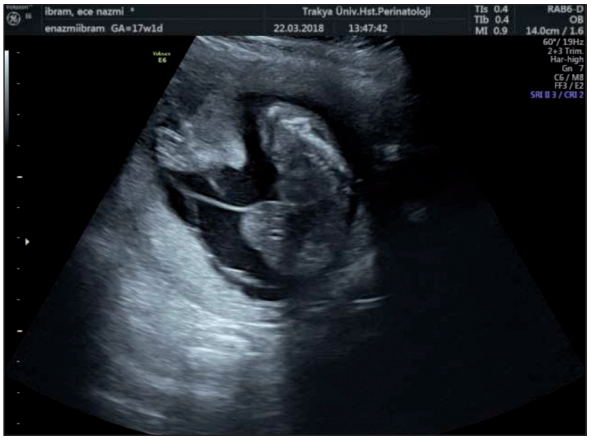

FIGURE 1: Ultrasonographic image of the fetus with multiple amniotic bands. Fetus with scoliosis with abdominal organs evisceration is seen in the photo.

and showed us single umbilical artery. Club foot also detected. No facial cleft or encephalocele detected with normal cloacal structures. Diagnosis of LBWC was made. We talked to the family about the poor prognosis and fatal outcome of the fetus. Family accepted termination of pregnancy. The informed consent was obtained from the patient. We put 600 mg misoprostol vaginally. Delivering of fetus happened after 16 hours (Figure 2, Figure 3). We did suction curettage for cavum control.

DISCUSSION

LBWC is also known as the body-stalk syndrome. It is a rarely seen clinical entity that is represented by exencephaly/encephalocele with or without craniofacial defect (56%), thoraco- and/or abdominoschisis, and limb defect (95%). There are some hypotheses to explain the pathogenesis of limb body wall complex. Some of them early amnion disruptions, embryonic dysplasia, and vascular disruption in early pregnancy.[3] Reported prevalence ranges from 0.4 to 3.2 per 100.000 live births.[4] Most fetuses are aborted, either spontaneously or by medical induction. Most of the remaining babies are stillborn, while postnatal survival for a significant duration is extremely rare. We did medical induction in our case. The diagnostic criteria for LBWC is still debatable. The commonly used one made by Van Allen et al. in1987 Russo et al. in 1993 identified two different phenotypes.[5] Type I (craniofacial defects, facial clefts, amniotic adhesions and amniotic band syndrome) and Type II (No craniofacial defects, Urogenital anomalies, imperforate anus, lumbosacral meningomyelocele, severe kyphoscolosis and placental anomalies). The sole criteria used by Russo et al. was craniofacial defects in Type I and many other thorasic and abdominal anomalies in Type II. Sahinoglu et al. in 2007 made a new classification: Type I (Fetus has craniofacial defect and intact thoracoabdominal wall, often normal placenta and umbilical cord but rarely attached to the malformated cranial structures), Type II (Fetus has supraumbilical, usually laterally located (often left side) large thoraco abdominal wall defect.[6] Eventrated abdominal organs enveloped within the amniotic sheet connects to the skin margin of the wall defect. No well-formed umbilical cord and usually normal cloacal structures are detected). Type III (Fetus has infraumbilical abdominal wall defect with intact thorax. The placenta is attached broadly to the skin at the site of defect. The abdominal organs are eventrated into the extraembryonic coelomic cavity. The cloacal structures are almost always malformed or absent). Our case obeyed Type II criteria.

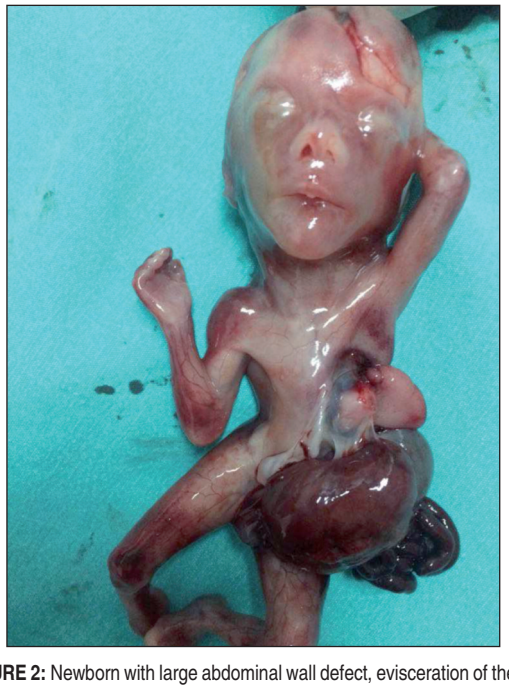

FIGURE 2: Newborn with large abdominal wall defect, evisceration of the abdominal organs, lower limbs with clubfoot.

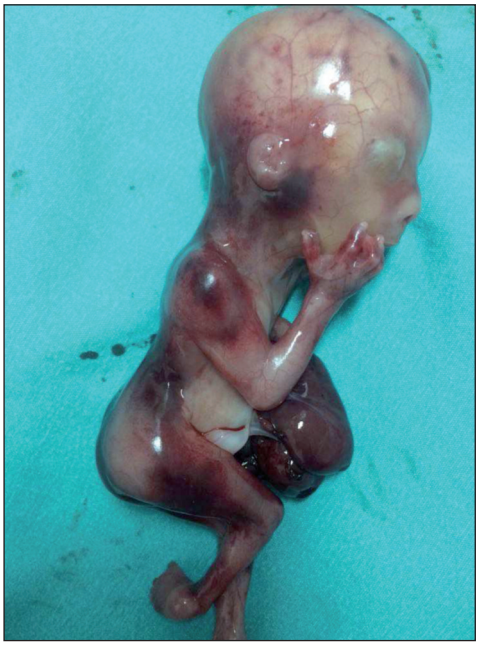

FIGURE 3: Transverse view of the fetus with abdominal organs evisserated and club foot is seen.

Bad outcome of LBWC makes it very necessary for an early prenatal diagnosis. Ultrasonographic detection of abdominoschisis, scoliosis, abnormalities of lower extremities, a single umbilical artery demonstrated by a color doppler, short umbilical cord and extremely elevated level of maternal serum alpha fetoprotein is the key to make early diagnosis.[7] Our patient didn't make combined or second trimester screening test. It is also important that LBWC should be differentiated from common abdominal wall defects such as gastroschisis, omphalocele and uncommon entities like ectopia cordis, amniotic band syndrome, cloacal dystrophy, and urachal cyst.[8,9] In our case there were many amniotic bands. Maybe a routine, detailed anomaly scan for all pregnant women in the second trimester by a trained and experienced perinatologists can help to minimize cases of misdiagnosis. Although the pathogenesis of this congenital condition with LBWC is not known currently, amniotic bands, vascular disruption or abnormal embryonic folding and sporadic genetic mutations can be counted among the underlying causes. It has also proposed some mutations in genes related to laterality and caudal development even though the recurrence risk is low.[10,11] There is no consensus about the etiology of LBWC even though the majority of cases of LBWC are sporadic, some women may have an underlying genetic predisposition.[12,13] We sent chromosomal analysis from fetus. Result was normal 46-XY karyotype. It is necessary to advise the patient not to use of alcohol, cigarettes and other drugs. It is seen in a younger age group and cocaine abuser. We should offer an ultrasound scan in a tertiary center during the subsequent pregnancy.

Informed Consent

The authors certify that they have obtained all appropriate patient consent forms.

Source of Finance

During this study, no financial or spiritual support was received neither from any pharmaceutical company that has a direct connection with the research subject, nor from a company that provides or produces medical instruments and materials which may negatively affect the evaluation process of this study.

Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Cem Yener;* ***Design:*** *Cem Yener;* ***Control/Supervision:*** *Füsun Varol;* ***Data Collection and/or Processing:*** *Işıl Uzun;* ***Analysis and/or Interpretation:*** *Cenk Sayın;* ***Literature Review:*** *Füsun Varol;* ***Writing the Article:*** *Cem Yener;* ***Critical Review:*** *Cenk Sayın;* ***References and Fundings:*** *Füsun Varol, Cenk Sayın;* ***Materials:*** *Cem Yener.*

REFERENCES

1. Patten RM, Val Allen M, Mack LA, Wilson D, Nyberg D, Hirsch J, et al. Limb-body wall complex: in utero sonographic diagnosis of a complicated fetal malformation. AJR Am J Roentgenol. 1986;146(5):1019-24. [Crossref]
2. Kocherla K, Kumari V, Kocherla PR. Prenatal diagnosis of body stalk complex: a rare entity and review of literature. Indian J Radiol Imaging. 2015;25(1):67-70. [Crossref] [PubMed] [PMC]
3. Kamudhamas A, Manusook S. Limb-body wall complex, report of 2 cases with their quintessence is prenatal diagnosis. J Med Assoc Thai. 2001;84(4):602-8. [PubMed]
4. Bohîlţea RE, Tufan CF, Cîrstoiu MM, Dumitru AV, Georgescu TA, Sajin M, et al. Body stalk anomaly in a monochorionic-diamniotic twin pregnancy-case report and review of the literature. Rom J Morphol Embryol. 2017;58(4):1453-60. [PubMed]
5. Russo R, D'Armiento M, Angrisani P, Vecchione R. Limb body wall complex: a critical review and a nosological proposal. Am J Med Genet. 1993;47(6):893-900. [Crossref] [PubMed]
6. Sahinoglu Z, Uludogan M, Arik H, Aydin A, Kucukbas M, Bilgic R, et al. Prenatal ultrasonographical features of limb body wall complex: a review of etiopathogenesis and a new classification. Fetal Pediatr Pathol. 2007;26(3):135-51. [Crossref] [PubMed]
7. Van Allen MI, Curry C, Walden CE, Gallagher L, Patten RM. Limb-body wall complex: II. Limb and spine defects. Am J Med Genet. 1987;28(3):549-56. [Crossref] [PubMed]
8. Torpin R. Amniochorionic mesoblastic fibrous strings and amnionic bands: associated constricting fetal malformations or fetal death. Am J Obstet Gynecol. 1965;91:65-75. [Crossref]
9. Zeidler S, Oudesluijs GG, Schoonderwaldt EM, Van Bever Y. Early prenatal disruption; a foetus with features of severe limb body wall sequence, body stalk anomaly and amniotic bands. Genet Couns. 2014;25(3):315-20. [PubMed]
10. Plakkal N, John J, Jacob SE, Chithira J, Sampath S. Limb body wall complex in a still born fetus: a case report. Cases J. 2008;1(1):86. [Crossref] [PubMed] [PMC]
11. Socolov D, Terinte C, Gorduza V, Socolov R, Puiu JM. Limb body wall complex-case presentation and literature review. Rom J Leg Med. 2009;2(2):133-8. [Crossref]
12. Bhat A, Ilyas M, Dev G. Prenatal sonographic diagnosis of limb-body wall complex: case series of a rare congenital anomaly. Radiol Case Rep. 2016;11(2):116-20. [Crossref] [PubMed] [PMC]
13. Gajzer DC, Hirzel AC, Saigal G, Rojas CP, Rodriguez MM. Possible genetic origin of limb-body wall complex. Fetal Pediatr Pathol. 2015;34(4):257-70. [Crossref] [PubMed] [PMC]