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Case Report

Prenatal diagnosis of limb body wall complex: early sonographic findings

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ABSTRACT

Limb body wall complex (LBWC) is a rare syndrome that consists of, combination of multiple fetal malformations of the thoracoabdominal wall, extremities, cranium, facial and spine. The sonographic hallmarks are abdominal/thoracic wall defect, neural tube defect, severe scoliosis, positional deformity and abnormal fetal membranes**.** Authors present a case of LBWC abnormality, which was detected in the antenatal ultrasonography at the POG of 13 weeks and later confirmed in the abortus. Early diagnosis of LBWC is only possible through ultrasonography, and it being a fatal condition needs to be distinguished from gastroschisis and omphalocele, which has a better prognosis.

Keywords: Limb body wall complex, Ultrasonography, Body stalk anomaly

INTRODUCTION

LBWC is a rare syndrome having a combination of multiple fetal malformations of the thoracoabdominal wall, extremities, cranium, facial and spine.[1] The sonographic hallmarks are abdominal/thoracic wall defect, neural tube defect, severe scoliosis, positional deformity and abnormal fetal membranes, and diagnosis can be established if 2 or 3 of these features are present.[2-4] Early diagnosis of LBWC is possible through ultrasonography, and it being a condition which is incompatible with life, therefore needs to be distinguished from gastroschisis and omphalocele, which have a relatively better prognosis. The grave prognosis of LBWC necessitates an early antenatal diagnosis and medical termination of the pregnancy. Authors present a case of LBWC abnormality, which was detected in the antenatal ultrasonography at the POG of 13 weeks and later confirmed in the abortus by necropsy.

CASE REPORT

A 26-year-old primigravida with a 3-month amenorrhea was referred to our department for routine ultrasonography and NT/NB scan. She had a normal blood profile with no history of hyperemesis or GDM. The ultrasound findings showed CRL measures 5.2 cm corresponding to 11 weeks 6 days.

There was a large anterior abdominal wall defect with herniation of abdominal contents (liver, spleen and bowel loops) into the extra amniotic space. There was the presence of Ectopia Cordis, where having heart is partially outside the thoracic cavity.

The spinal deformity having altered curvature in the form of scoliosis of the thoracolumbar spine was also seen. No overt encephalocele or cranial abnormalities detected. The nuchal translucency (NT) was measured at 1.2 mm, within normal limits, and nasal bones were present: the ductus venosus wave was normal.

The female was referred back to the Department of Gynaecology and Obstetrics for further management with a diagnosis of limb body wall complex. On counselling, the patient about the fatal outcome of the anomaly, the pregnancy was terminated, and ultrasonography findings confirmed on necropsy (Figure 1-4).

Table 1: Differentials of LBWC anomaly.

Characteristic	LBWC	Gastroschisis	Omphalocele
Timing of diagnosis	End of first trimester	Second trimester	Second trimester
Location of defect	Thoracoabdominal wall, large	Paraumbilical, right sided	Midline, abdomen
Other associated anomalies	Multiple severe (Neural tube defect, severe scoliosis, positional deformity, abnormal fetal membranes etc)	Usually, isolated defect	May be associated with other anomalies, chromosomal anomalies
Umbilical cord	Short or absent	normal	Inserts into abnormal sac
Spinal	Common, severe	Absent	Rare
Prognosis	fatal	Good with surgical repair	Variables depending on associated anomalies
AFP levels	Raised	Raised	Normal or mildly elevated

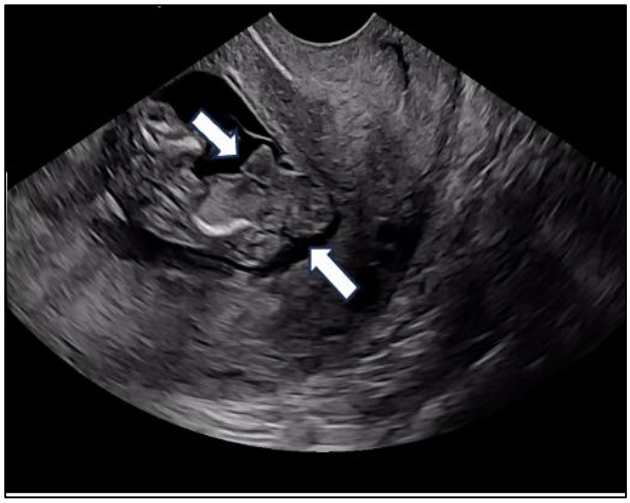

Figure 1: A large ventral abdominal wall defect through which gut loops (arrow) are seen herniating into the extra-amniotic space.

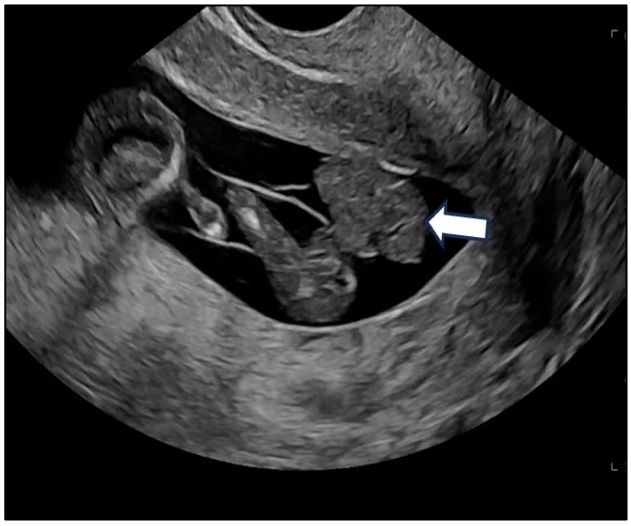

Figure 2: Herniation of the liver (arrow) into the extra-amniotic space through the ventral abdominal wall defect.

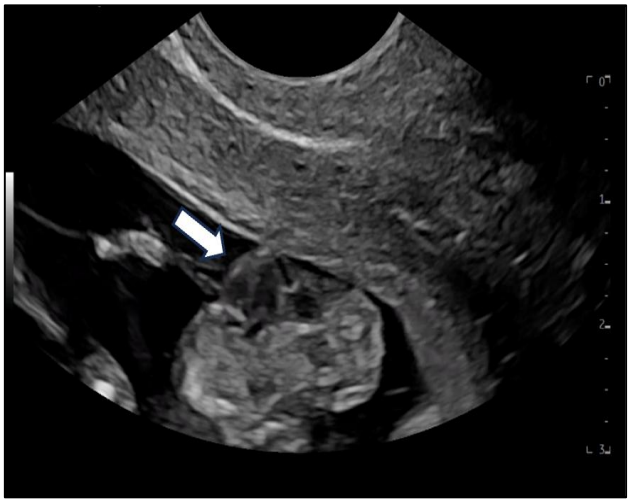

Figure 3: Ectopia cordis (arrow) in which the heart is present partially outside the thoracic cavity.

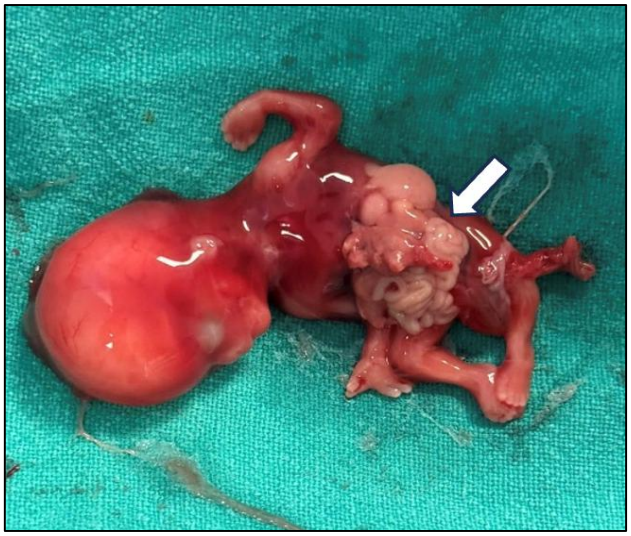

Figure 4: Post-termination necropsy specimen showing a large ventral abdominal wall defect through which abdominal organs (arrow) are seen herniating.

DISCUSSION

LBWC is a fatal, multiple fetal malformation syndrome that consists of a wide spectrum of anomalies in the body wall.[1] This entity was described for the first time by Van Allen et al in 1987, with diagnostic criteria being two out of three of the following: exencephaly/encephalocele with facial clefts, thoracic and or abdominochisis and limb defect.[2,3] The aetiology of LBWC is not clear; the hypothesis proposed by Van Allen and Tropin fails to explain multiple anomalies of this entity. The vascular disruption theory proposed by Van Allen et al suggests that congenital malformation arises from disrupted embryonic blood supply, which is supported by animal studies involving amniotic puncture and vessel ligation. However, human cases often lack immediate vascular compromise post-rupture, thus weakening the theory; moreover, it fails to even explain the multiple anomalies. Tropin's amniotic band theory suggests fetal malformations occur when amniotic rupture causes fibrous bands to entrap fetal parts, with severity depending upon timing. There are inconsistencies in this theory, especially the high prevalence of internal anomalies, which are left unexplained. The most accepted theory is early embryonal dysplasia by Harwig et al given in 1989, which attributed the defect to abnormal embryonic folding due to ectodermal placode malfunction. It states that the vascular disruption occurs secondary to local vessel hypoplasia rather than being a primary cause.

The diagnosis of LBWC can be established by raised maternal alpha-fetoprotein levels. The ultrasonographic findings at the end of first trimester are abdominal wall defect. Severe scoliosis or kyphosis of the spine, short umbilical cord and limb abnormalities. Color doppler flow imaging shows one artery and one vein.[4] This condition is fatal and needs to be distinguished from gastroschisis and omphalocele, which have a relatively better prognosis.[5] The gastroschisis is an isolated paraumbilical defect, whereas an omphalocele is a midline defect and may pose a diagnostic challenge as it is sometimes associated with other anomalies. Various differential diagnoses are shown in Table 1. The preferred treatment method is the earliest possible diagnosis and termination of the pregnancy under medical supervision, and doing the necropsy, which is the gold standard for the final diagnosis.[6-8]

CONCLUSION

Early diagnosis of limb body wall complex anomalies is achievable through prenatal ultrasonography, which facilitates the timely identification of their characteristic features. Given its uniformly fatal prognosis, it is crucial to differentiate LBWC from anterior abdominal wall defects such as gastroschisis and omphalocele, which have better survival outcomes. Published literature underscores the role of high-resolution ultrasound in enabling this distinction during early gestation.

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