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Case Report

Prenatal sonographic diagnosis of limb-body wall complex: case series of a rare congenital anomaly

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ABSTRACT

Three case reports of a rare congenital anomaly “limb-body wall complex” also known as “body stalk syndrome” are presented with prenatal ultrasonographic diagnostic features, immediate after delivery evaluation, and histopathologic analysis.

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Introduction

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

- (1) Exencephaly and/or encephalocele with facial clefts,
- (2) Thoraco and/or abdominoschisis, and
- (3) Limb defects [1].

The sonographic hallmarks of LBWC are neural-tube abnormalities, severe scoliosis, positional deformities, and abnormalities of fetal membranes [2]. 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.” Unfortunately, there is no cure for LBWC, and it is generally considered to be incompatible with life (fatal) [3]. The poor prognosis of LBWC necessitates an early antenatal diagnosis and termination of pregnancy.

Case reports

Case report 1

A 26-year-old primigravida female with a 7-month amenorrhea was referred to our department for routine ultrasonography and fetal well-being study. She had normal blood

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profile and O +ve blood group. Hemoglobin was 10.5gm/dL. The ultrasonography revealed normal head with biparietal diameter (BPD) corresponding to 26 weeks 3 days. However, a large abdominal wall defect, with liver and gut coils herniating through it, into the liquor amnii was seen. The herniated organs formed an entangled complex covered by fetal membranes. Spinal dysraphism along with a large meningocele was also visualized in the lumbosacral region (Fig. 1A). Further study revealed bilateral clubfoot deformity in the fetus (Fig. 1B). Color Doppler study showed single umbilical artery supplying the liver (Fig. 1C). No anomaly was detected in the eyes, palate, lips, face, and thoracic region. The female was referred back to department of Gynecology and Obstetrics for further management with diagnosis of LBWC.

On counseling, the patient about the fatal outcome of the anomaly, the pregnancy was terminated and ultrasonography findings confirmed thereof (Fig. 1D and E). Histopathologic analysis of the umbilical cord revealed single umbilical artery (Fig. 1F) and hypoplastic right internal iliac artery.

Case report 2

A 22-year-old G₂P₁ female with a 6-month amenorrhea was referred for fetal ultrasonography to the Radiology department. The blood profile revealed decreased Hemoglobin (8.5%), A +ve blood group and normal coagulation profile. The ultrasonography revealed gestational age of 22 weeks using BPD. Furthermore, it revealed a large abdominal wall defect

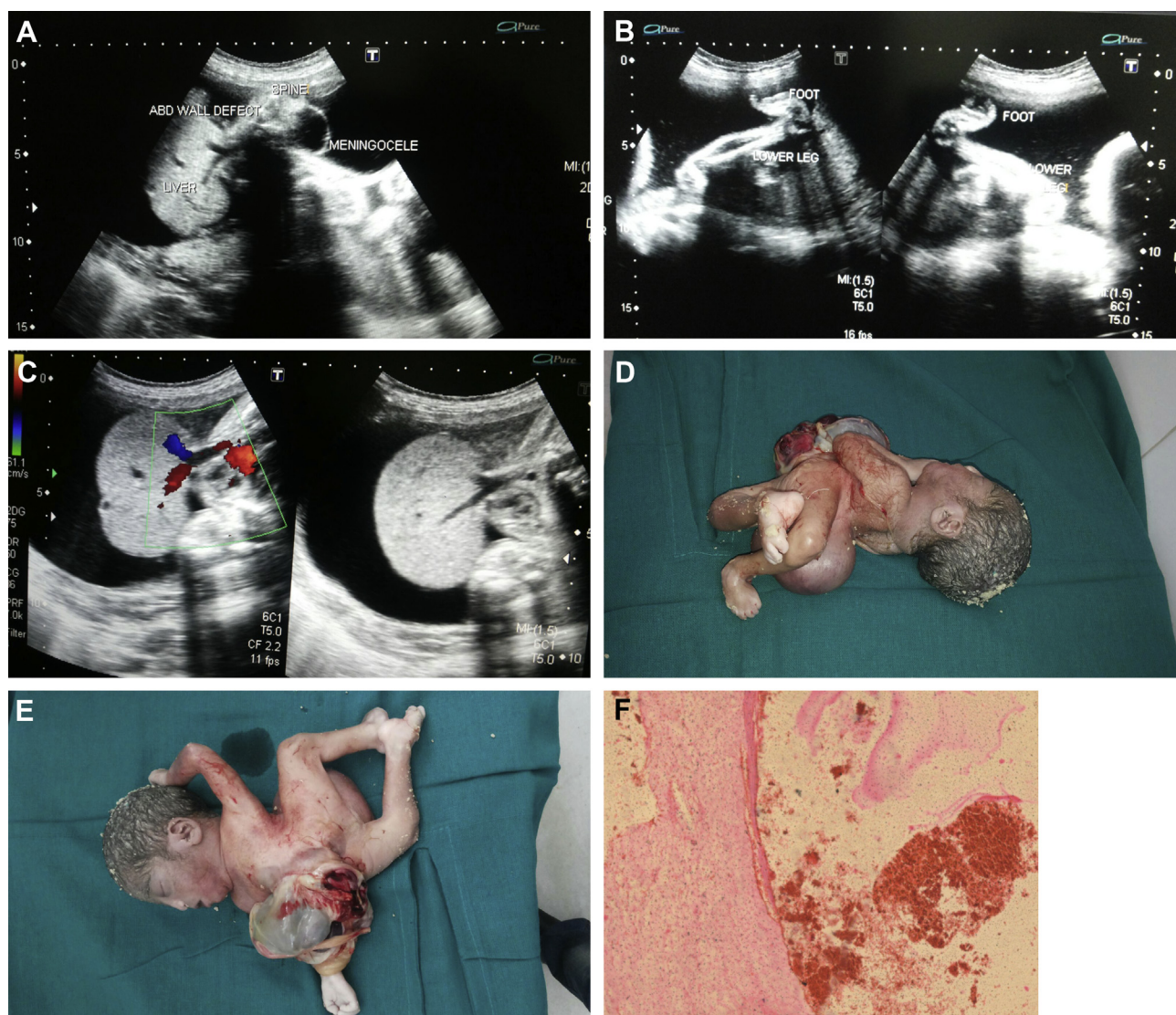


Fig. 1 – (A) Longitudinal view of ultrasound image showing the large abdominal wall defect with liver and gut-coils herniating through it into the liquor amnii. Also depicted is the meningocele in lumbosacral region and spinal dysraphism. (B) USG images demonstrating bilateral clubfeet deformity. (C) Color Doppler image showing the blood flow into liver of the fetus via the single umbilical artery. (D) Immediate post delivery image showing the large meningocele, herniated abdominal contents through abdominal wall defect, spinal dysraphism and club foot deformity is also seen confirming USG findings. (E) Post delivery image showing the large abdominal defect through which abdominal organs are seen herniating. Sex of the fetus is not distinguished. (F) Histopathology slide showing single umbilical artery with RBC's in the lumen.

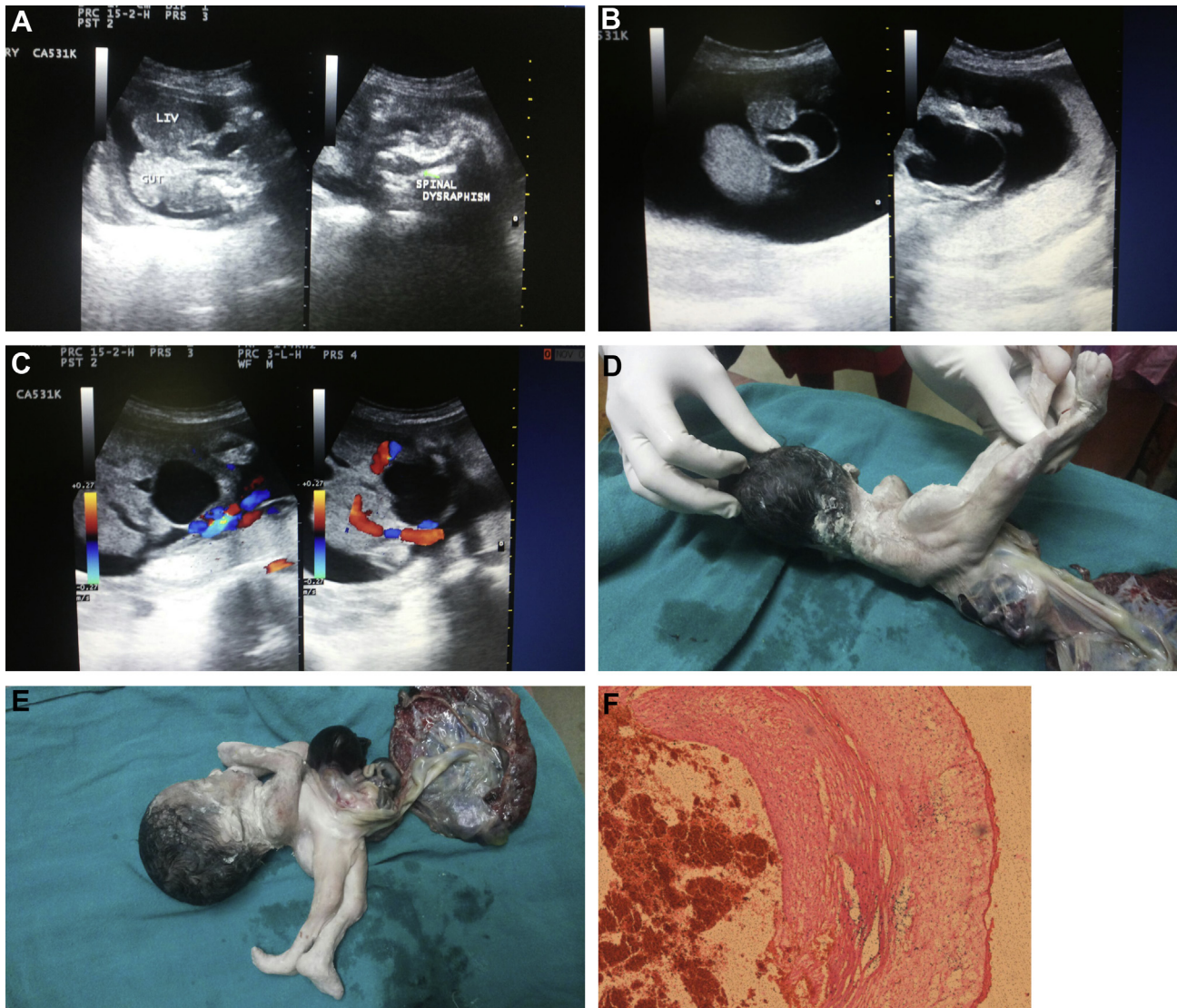


Fig. 2 – (A) Longitudinal USG view showing spinal dysraphism (left window), herniated gut contents and herniated liver (right window). **(B)** USG view demonstrating large meningocele and herniated gut contents and liver. **(C)** Color Doppler image showing blood flow into the single umbilical artery. **(D)** Immediate post delivery image showing placenta with short umbilical cord, large meningocele, spinal dysraphism and herniated abdominal contents. **(E)** Post delivery image showing large defect in the anterior abdominal wall defect with liver and gut contents herniating through it. A part of meningocele is also visible. **(F)** Histopathology slide showing single umbilical artery with RBC's in the lumen.

with gut coils and liver seen herniating into the amniotic cavity. Spinal dysraphism was also seen in this fetus (Fig. 2A). Also, a large meningocele along with entangled mass of herniated contents with the fetal membranes covering them was seen (Fig. 2B). Color Doppler study was performed and revealed single umbilical artery (Fig. 2C). No anomaly was seen in the head and thoracic region. The diagnosis of LBWC was made. The female was counseled about the poor prognosis and fatal outcome of the fetus and pregnancy was terminated by the obstetrician. The findings were confirmed after delivery by the obstetrician (Fig. 2D and E). Histopathology confirmed the single umbilical artery (Fig. 2F).

Case report 3

A 23-year-old G₃P₂ female with a 6 and half-month amenorrhea was evaluated for congenital anomalies by ultrasonography. The gestational age was 24 weeks by BPD. The ultrasonography revealed a large abdominal wall defect with abdominal contents seen herniating through it with meningocele (Fig. 3A and B). No other anomaly was detected. The diagnosis of LBWC was given. Color Doppler study could not be performed because of some technical reasons. The female was counseled and the pregnancy terminated and the findings confirmed thereof (Fig. 3C and D). Histopathology did not reveal any significant vascular anomaly.

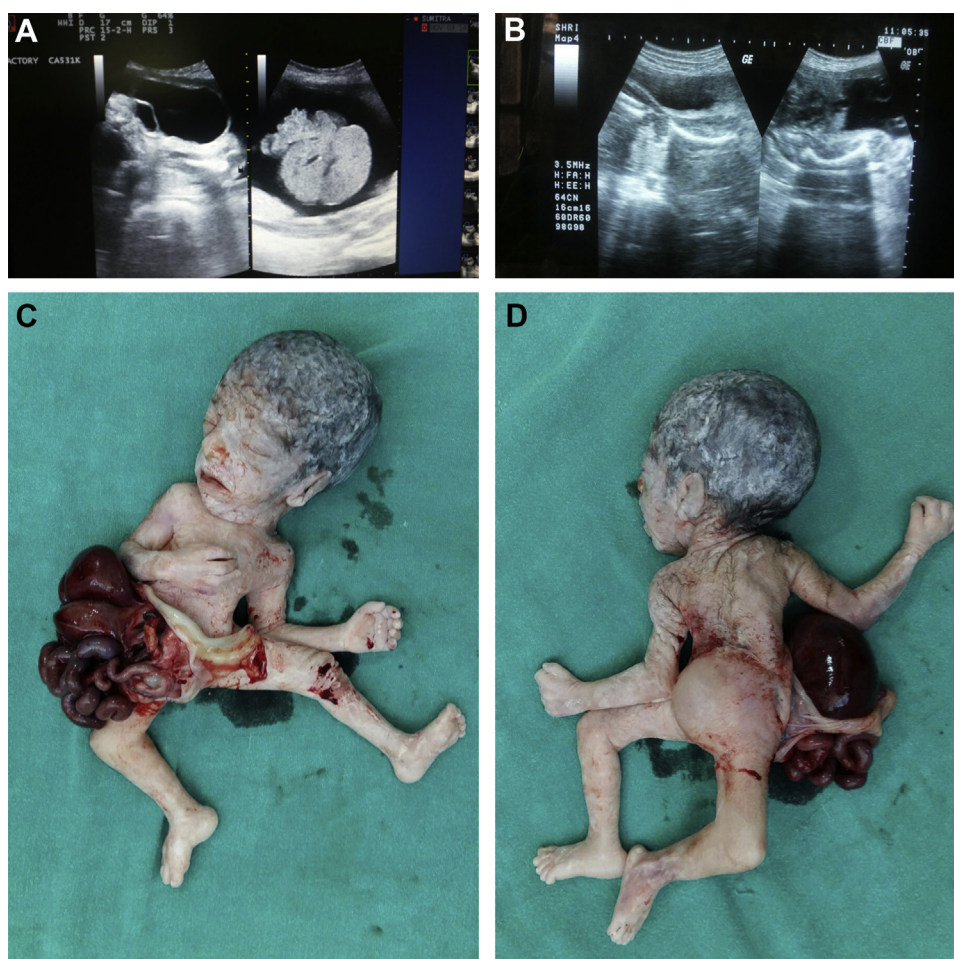


Fig. 3 – (A) Longitudinal USG view showing the herniated abdominal contents and meningocele. (B) USG view demonstrating fetal leg and a part of herniated contents. (C) Post delivery image showing large abdominal defect and contents herniating through it. (D) Immediate post delivery image showing meningocele and herniated abdominal contents.

Discussion

The incidence of LBWC at birth is about 0.32 per 100,000 births because majority of affected fetuses undergo intrauterine deaths [4]. Traditionally, the diagnosis has been based on the Van Allen et al criteria, that is, the presence of any 2 of the 3 following anomalies:

- (1) Exencephaly or encephalocele with facial clefts,
- (2) Thoraco and/or abdominoschisis, and
- (3) Limb defects.

The exact etiology of this condition is still unclear, Tropin's amniotic band theory, and Van Allen's vascular theory failed to explain all the anomalies in LBWC [5].

Tropin suggested that once the amnion is ruptured, the fetus lies outside the amniotic cavity and bands extending from the chorionic side of the cavity entrap various parts of the fetus and disturb normal development. The severity depends on the period of gestation during which the bands develop, especially when accompanied with

oligohydramnios. Early rupture would lead to more severe malformations (eg, craniofacial and visceral), whereas late rupture would lead to milder forms. Higginbottom, in 1979, described 79 patients that supported the band theory resulting from observations of unusual facial clefts, which were not along the planes of facial closure. Also in support of the band theory Bhat, in his case report, described the presence of a well-formed amputated distal portion of 1 leg and fibrous bands coiling around the fingers of the amputated segment. Many investigators have questioned this theory. Van Allen noted bands only in 40% of cases and internal defects in 95% of cases; features that could not be attributed to amniotic bands. Herva has refuted this theory on the basis of the high prevalence of internal anomalies. Further criticism of the amniotic disruption theory is derived from the reports of Yang who described that antenatal amniotic rupture results in extra-amniotic pregnancy that displays no evidence of amniotic bands. Anomalies not explainable by the amniotic disruption theory are internal malformations—tetralogy of Fallot, rudimentary upper limb, abnormal number and morphology of umbilical vessels and short great toe.

Vascular disruption theory (VDT) was proposed by Van Allen and implies that various congenital malformations may generally result from vascular disruption. Vascular disruption is described as events that negatively influence normal embryonic blood supply during embryogenesis, thereby interrupting normal morphogenesis, or destroying previously existing structures. VDT is supported by experimental animal studies. Puncturing the amniotic cavity or ligating umbilical blood vessels have produced LBWC. In amnion puncture experiments, up to one-third of the total amniotic fluid was withdrawn, thereby greatly influencing fetoplacental circulation. Many investigators have also questioned this theory. In human, no immediate loss of amniotic fluid is expected after rupture of the amniotic membrane during early pregnancy (because the chorion is usually spared), so no such vascular change is expected. However, when adhesion and bands form, this factor occurs and plays a role in disruption. Anomalies not explainable with VDT are anencephaly, absent orbit (one sided), oblique facial cleft, deformed nose (single nostril), rocker bottom feet, pulmonary hypoplasia, unlobulated liver, abnormal number of umbilical vessels, cardiac defect (Tetralogy of Fallot), skin tag, and facial dysmorphism. Hence, this pathogenetic mechanism cannot explain all the anomalies.

The most accepted theory is early embryonal dysplasia put forward by Hartwig et al [6], in 1989. According to this, there will be an abnormal embryonic folding related to malfunctioning of the body wall ectodermal placode. This leads to defective closure of embryonic abdominal wall umbilical abnormality and persistence of extraembryonic coelom communicating with the abdominal cavity. Some suggest that vascular disruption is secondary to hypoplasia of the blood vessels in the affected area rather than being primary etiologic factor [7].

This anomaly does not have any sex predilection and recurrence of this condition was observed in 2 families suggesting a possible genetic etiology [8]. The diagnosis of this condition can be established by measuring maternal serum alpha fetoprotein levels. Prenatal ultrasound examination can detect this anomaly as early as first trimester (usually by the end of first trimester) [9].

In our cases, the criteria laid by Van Allen were applied, and it fitted in all the cases for diagnosis of LBWC. Although vascular abnormality was seen in 2 cases and the third case revealed no vascular abnormality, our consensus remains that vascular maldevelopment is the most probable cause for the pathogenesis of LBWC.

Learning points

- (1) LBWC is a rare congenital anomaly with no sex predilection, and approximately 250 cases are reported in the literature till date.
- (2) It has a possible genetic cause, but the most accepted theory is “early embryonal dysplasia.”
- (3) Once diagnosed antenatally, pregnancy should be terminated as it is incompatible with life.
- (4) It is important to differentiate them from other anterior abdominal wall defects such as gastroschisis and omphalocele as these have a more favorable prognosis as compared with LBWC.
- (5) If combination of omphalocele and scoliosis is seen in a fetus, it should raise suspicion of LBWC.
- (6) Sonographic hallmarks of LBWC thoraco and/or abdominoschisis, neural tube defects, severe scoliosis, positional deformities, and abnormality of fetal membranes, should be kept in mind if a suspicion of LBWC is there.

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