Title: Limb-Body Wall Complex (LBWC): A Case Report and Review of Literature

Abstract

Objective

Limb-Body Wall Complex (LBWC) is a rare and severe congenital anomaly characterized by malformations of the limbs, body wall, and internal organs. This case report presents a clinical diagnosis of LBWC, reviews the current literature, and highlights the challenges in diagnosis and management of this condition.

Case Presentation

A 21-year-old pregnant woman with no prenatal follow-up was admitted to our obstetric emergency unit at 36 weeks and 6 days of gestation for imminent delivery in breech presentation. A cesarean section was performed, and macroscopic examination revealed a male newborn with severe malformations, including absence of the abdominal wall with organ protrusion, placento-abdominal attachment with a short umbilical cord, thoracic narrowing, scoliosis, and limb deformities. Despite visible cardiac activity, the neonate died shortly after birth due to severe multisystemic defects.

Conclusion

LBWC is a rare and lethal condition with no curative treatment. Early antenatal diagnosis is essential for appropriate counseling and palliative care. The underlying etiology remains unclear, with embryological and vascular disruption theories being the most cited. This case underscores the importance of recognizing LBWC early and providing comprehensive support to affected families.

Keywords: Limb-Body Wall Complex, congenital anomaly, antenatal diagnosis, macroscopic findings, palliative care.

Introduction

Limb-Body Wall Complex (LBWC) is a rare and severe congenital disorder characterized by malformations of the limbs, body wall, and internal organs, often leading to life-threatening complications and poor outcomes [1]. The condition is typically diagnosed based on the criteria established by Van Allen et al., which require at least two of the following abnormalities: (1) exencephaly or encephalocele with facial clefts, (2) thoraco-abdominal wall defects, or (3) limb deformities [2].

Although prenatal ultrasound can aid in identifying LBWC, the condition is frequently underdiagnosed, even postnatally, and is often misclassified as a "polymalformative syndrome" [3]. Early diagnosis is crucial for parental counseling and management planning, particularly in resource-limited settings where access to advanced imaging techniques may be restricted [4].

In this report, we present a case of LBWC suspected on antenatal ultrasound and confirmed clinically after delivery. The patient, a 21-year-old woman with no prenatal follow-up, was admitted to our obstetric emergency unit at 36 weeks and 6 days of gestation for imminent breech delivery. This case highlights the diagnostic challenges and management implications of LBWC, emphasizing the importance of early recognition and comprehensive care. Our work has been reported in accordance with the SCARE guidelines [5].

Case Presentation

A 21-year-old pregnant woman, gravida 3 para 1 (G3P1), with a history of two miscarriages and no family history of congenital malformations or consanguinity, presented to our obstetric emergency unit at 36 weeks and 6 days of gestation for imminent delivery. She had no prior prenatal care and no history of teratogen exposure, hypertension, diabetes, or use of drugs or medicinal plants during pregnancy.

Upon admission, the patient was stable and in good general condition. Obstetric examination revealed a uterine height appropriate for gestational age, a well-perceived fetal heart rate, and decreased fetal movements. Vaginal examination confirmed ruptured membranes, an incomplete breech presentation, and cervical dilation of 3 cm, indicating active labor.

An obstetrical ultrasound showed a singleton pregnancy with a fundal placenta, normal amniotic fluid volume, and a polymalformed fetus in breech presentation . The findings included severe deformities of both lower limbs, complete absence of the abdominal wall leading to protrusion of the heart, liver, spleen, and other organs, as well as placento-abdominal adhesions with thoracic narrowing. These features strongly suggest a complex congenital malformation, most likely Limb-Body Wall Complex (LBWC) (Figures 1-2).

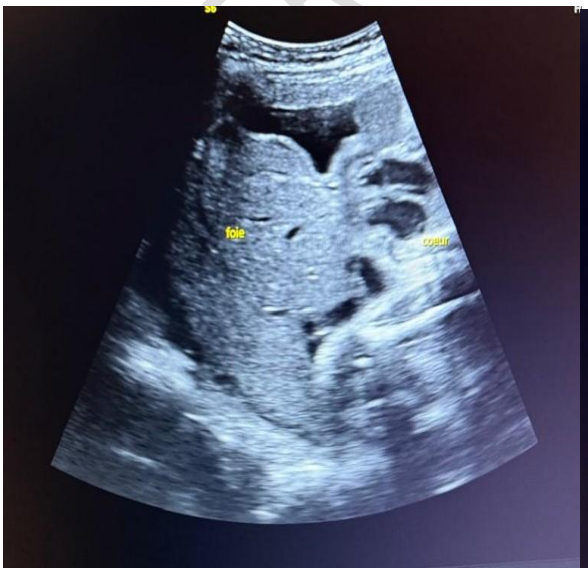

Figure 1 : "Prenatal ultrasound of ectopia cordis

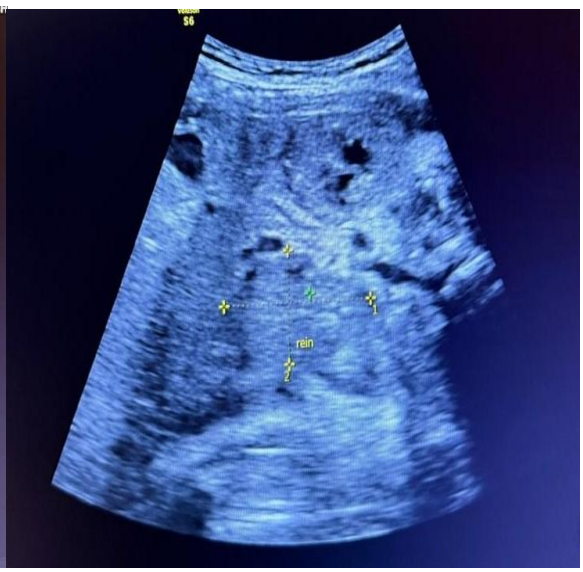

Figure 2 : Evisceration of the visceraon ultrasound

The patient was thoroughly informed about the polymalformed fetus and the urgent need for a cesarean section. After obtaining her informed consent, she was transferred to the operating room. A cesarean section was performed, and a male neonate was delivered. Macroscopic examination of the newborn revealed severe malformations, including:

- Deformity of both lower limbs (Figure 3).
- Absence of the abdominal wall with protrusion of the heart, liver, spleen, small intestine, and colon (Figure 4).
- Placento-abdominal attachment with a short umbilical cord.
- Thoracic narrowing and severe scoliosis (Figure 5).

Figure 3 : ***Limb-Body Wall Complex with Limbs malformations***.

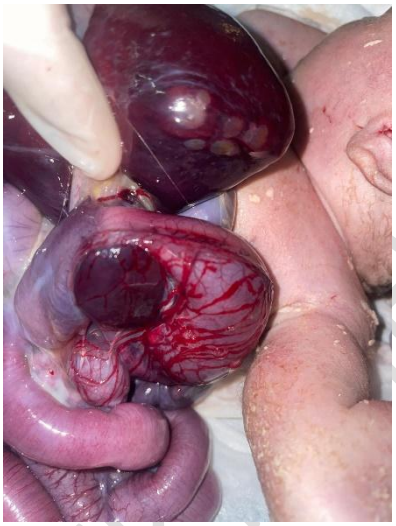

Figure 4 : ***Evisceration of stomach , liver, intestines ,spleen*** .

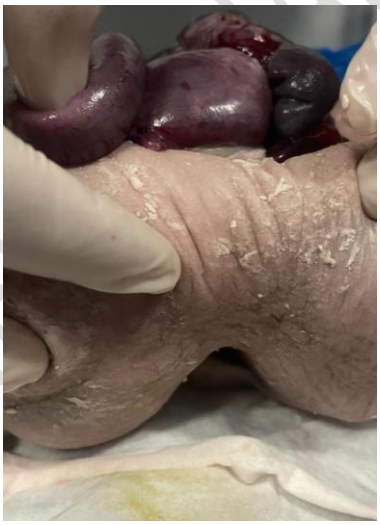

Figure 5 : ***severe scoliosis***.

The Apgar score was 02/10 at 1 minute and 01/10 at 5 minutes, with a birth weight of 2200g. Despite visible cardiac activity, the neonate died shortly after birth due to severe multisystemic defects. A skeletal radiograph could not be performed due to the emergency context, and anatomopathological examination was unavailable due to the absence of a fetopathology unit. However, the macroscopic findings were sufficient to confirm the diagnosis of Limb-Body Wall Complex (LBWC).

Discussion

This case report describes a male neonate with Limb-Body Wall Complex (LBWC), born to a 21-year-old woman with no prenatal follow-up. The newborn exhibited severe malformations, including abdominal wall defects, visceral protrusion, thoracic narrowing, scoliosis, and limb deformities, consistent with the diagnostic criteria established by Van Allen et al. [2]. Despite

visible cardiac activity at birth, the neonate died shortly after delivery due to multisystemic defects, underscoring the lethal nature of LBWC.

LBWC is a rare congenital anomaly, with an estimated incidence of 1 in 14,000 to 1 in 42,000 live births [6]. The condition is characterized by body wall defects, limb abnormalities, and internal organ malformations, often leading to stillbirth or early neonatal death. The absence of prenatal care in this case delayed diagnosis until delivery, highlighting the challenges of managing LBWC in resource-limited settings. Early prenatal diagnosis, particularly through ultrasound, is crucial for identifying characteristic findings such as abdominal wall defects, limb deformities, and thoracic narrowing [7]. In cases where ultrasound findings are inconclusive, prenatal MRI can provide additional diagnostic clarity, offering superior soft tissue contrast and detailed visualization of complex anomalies [8]. However, in resource-limited settings, access to MRI may be restricted, making ultrasound the primary diagnostic tool.

The exact etiology of LBWC remains unclear, with vascular disruption and amnion rupture being the most cited theories [9]. While these theories provide plausible explanations, neither fully accounts for the variability of LBWC cases. Our case supports the notion that LBWC results from a combination of genetic, vascular, and environmental factors. However, further research is needed to elucidate the underlying mechanisms and improve early detection.

There is currently no cure for LBWC. Therapeutic management focuses on symptom management, palliative care, and family support. In cases where survival is possible, surgical interventions may be considered to address specific defects, but the prognosis remains very guarded. Advances in genetic medicine and surgery may potentially offer new avenues in the future, but at present, the focus remains on improving the quality of life for affected neonates and providing comprehensive support to their families.

Early prenatal diagnosis is essential for offering families informed choices, including the possibility of medical termination of pregnancy (MTP) in cases of severe anomalies, where permitted by local laws. For instance, in many countries, MTP is allowed up to 24 weeks of gestation for lethal or severely disabling conditions. In our case, the absence of prenatal diagnosis eliminated this option, leaving the family unprepared for the tragic outcome.

Our findings align with previous reports of LBWC, such as those by Russo et al. [10], which describe similar cases with abdominal wall defects and limb deformities. However, unlike some rare cases where surgical interventions prolonged survival, our patient succumbed shortly after birth due to the severity of the anomalies. This variability in outcomes highlights the need for individualized care plans based on the specific malformations present.

This case report has several limitations, including the lack of prenatal follow-up, the absence of histopathological or radiographic confirmation, and the unavailability of genetic testing. Despite these limitations, the macroscopic findings were sufficient to confirm the diagnosis of LBWC.

Conclusion

Limb-Body Wall Complex (LBWC) is a rare and lethal congenital anomaly with no curative treatment. This case highlights the importance of early prenatal diagnosis for appropriate counseling and palliative care. The absence of prenatal follow-up in this instance delayed diagnosis until delivery, emphasizing the need for improved access to antenatal screening, particularly in resource-limited settings. Further research is needed to better understand the etiology of LBWC and explore potential therapeutic options.

Références

1. Bergman JEH, et al. Amniotic band syndrome and limb body wall complex in Europe 1980–2019. *Am J Med Genet A*. 2023.
2. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. *Am J Med Genet*. 1987;28(2):529-548. doi:10.1002/ajmg.1320280234.
3. D'Souza J, et al. Limb Body Wall Complex. *Med J Armed Forces India*. 2004.
4. Aguirre-Pascual E, et al. Prenatal MRI evaluation of limb-body wall complex. *Pediatr Radiol*. 2014.
5. Agha RA, et al. The SCARE Guidelines: Consensus-based surgical case report guidelines. *Int J Surg*. 2020.
6. Martínez-Frías ML, Bermejo E, Rodríguez-Pinilla E, Prieto L. Limb-body wall complex: Analysis of its relationship with amniotic bands using case reports. *Am J Med Genet*. 2000;91(3):153-157. doi:10.1002/(sici)1096-8628(20000320)91:3<153::aid-ajmg1>3.0.co;2-2.
7. Aguirre-Pascual E, Epeldegui-Torre T, Uriz-Moreno MJ, et al. Prenatal MRI evaluation of limb-body wall complex. *Pediatr Radiol*. 2014;44(11):1422-1429. doi:10.1007/s00247-014-3004-2.
8. Daskalakis G, Anastasakis E, Papantoniou N, et al. Limb-body wall complex: A case series and review of the literature. *J Matern Fetal Neonatal Med*. 2012;25(12):2612-2616. doi:10.3109/14767058.2012.704447.
9. Russo R, D'Armiento M, Vecchione R. Limb-body wall complex: A critical review and a nosological proposal. *Am J Med Genet*. 1993;47(6):893-900. doi:10.1002/ajmg.1320470617.
10. Russo R, D'Armiento M, Vecchione R. Limb-body wall complex: A critical review and a nosological proposal. *Am J Med Genet*. 1993;47(6):893-900. doi:10.1002/ajmg.1320470617.