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RESEARCH ARTICLE

THE "LIMB BODY WALL COMPLEX": ABOUT TWO CASES - A RARE ANOMALY

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Abstract

Limb Body Wall Complex (LBWC) is a rare and severe congenital anomaly characterized by multiple malformations, including defects in the anterior body wall, internal viscera, spine, central nervous system, and limbs. This article presents two prenatal cases of LBWC diagnosed at 15 and 31 weeks of gestation. Both cases highlight key phenotypic variations, including ectopiacordis, thoracoabdominoschisis, and limb anomalies, with differing placental attachment patterns. Diagnostic considerations, including ultrasound and maternal serum markers, are discussed alongside insights into etiopathogenesis and the uniformly poor prognosis. These cases underscore the importance of early prenatal diagnosis to differentiate LBWC from other anomalies and guide appropriate management, typically medical termination of pregnancy due to the fatal nature of the condition.

Introduction:-

Limb Body Wall Complex (LBWC) is a rare and severe polymalformative syndrome that was relatively obscure to obstetricians and sonographers until 1987, when Van Allen et al. provided a detailed description [1]. This condition is characterized by profound malformations involving the anterior body wall, internal viscera, central nervous system, spine, and limbs [2]. LBWC is exceptionally rare, with an estimated incidence of 1 in 14,000 to 1 in 31,000 pregnancies [26], and it is frequently associated with intrauterine fetal demise.

The diagnosis of LBWC is traditionally based on the criteria established by Van Allen et al., which require the presence of at least two out of three anomalies:

1. Exencephaly or encephalocele with facial clefts,
2. Thoracoabdominoschisis, and/or
3. Limb anomalies.

In this article, we present two cases of prenatal diagnosis of LBWC, identified at 15 weeks of gestation in a primigravida and at 31 weeks in a multigravida. These cases provide an opportunity to explore the diagnostic criteria, etiopathogenesis, and prognosis of this rare syndrome.

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Observation 1

The first case involved a 19-year-old primigravida (G1P0) who presented to the Obstetrics and Gynecology Department at the Mohammed VI University Hospital in Marrakech at 15 weeks of amenorrhea. She was referred by a private practitioner for the evaluation and management of suspected fetal malformations. The patient had no notable medical or obstetric history, including hypertension, diabetes, consanguinity, substance use, or medication use during pregnancy. On examination, she was hemodynamically and respiratorily stable, with unremarkable general and gynecological findings.

An obstetric ultrasound confirmed a singleton pregnancy complicated by a polymalformative syndrome, including ectopiacordis, omphalocele, and laparoschisis with exposed intestines and liver. Routine hematological and biochemical tests were within normal limits. Given the severity of the malformations, medical termination of pregnancy was performed.

Post-delivery macroscopic examination revealed a male fetus weighing 400 g. The fetus exhibited exencephaly, an asymmetric cleft lip, a pronounced anterior coelosomy with externalized liver and intestines, and thoracoschisis. Notably, no limb anomalies were observed **(Figure 1,2).** The fetus was attached to the placenta through both placento-cranial and placento-abdominal connections.

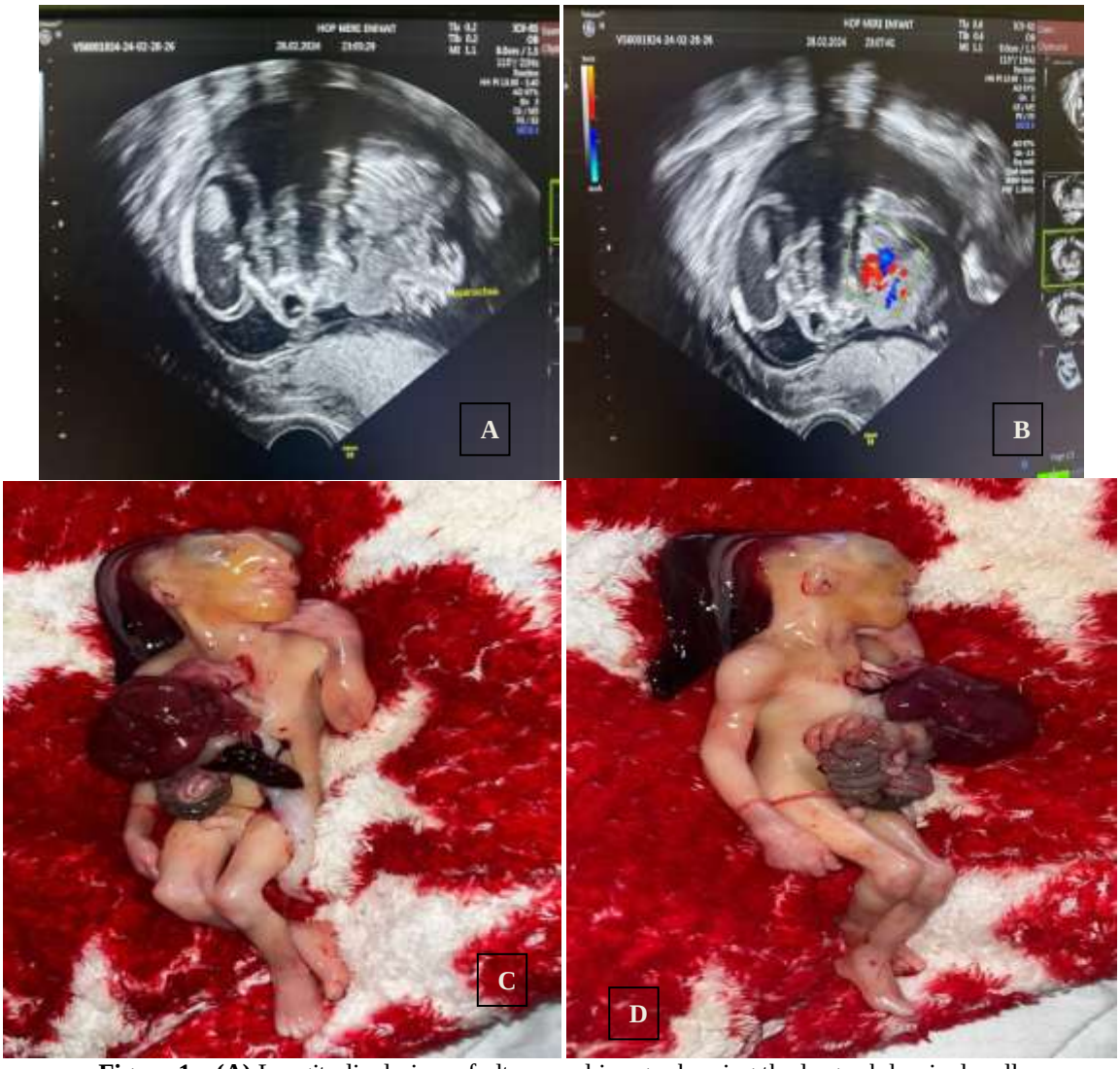

Figure 1:- (A) Longitudinal view of ultrasound image showing the large abdominal wall defect with liver and gut-coils herniating through it into the amniotic fluid, large meningocele

(B) Color Doppler image showing the blood in the ectopia cardis.
Figure 2: (C,D) Immediate post delivery image showing the large meningocele, herniated abdominal contents through abdominal wall defect, ectopiacardis, and a cleft lip.

Observation 2
The second case involved a 37-year-old multiparous patient (G4P3), O Rh-negative, with a history of two live births via vaginal delivery, a miscarriage at three months of gestation, and poorly controlled gestational diabetes. She presented to the emergency department of the Mohammed VI University Hospital at 31 weeks of amenorrhea due to premature rupture of membranes. The patient was referred from the provincial hospital in Tiznit for the evaluation and management of a polymalformative syndrome identified during pregnancy.

The patient denied any history of medication use during pregnancy, consanguinity, or substance use. She was hemodynamically and respiratorily stable, with an unremarkable general examination. Gynecological examination confirmed premature rupture of membranes without active labor.

Obstetric ultrasound revealed a singleton pregnancy complicated by a polymalformative syndrome, including ectopiacordis, laparoschisis with exposed intestines and liver, lower limb abnormalities (clubfoot), and absence of the left upper limb. No cerebral anomalies were identified (Figures G, H). Routine hematological and biochemical investigations were normal.
The patient underwent medical termination of pregnancy. Post-delivery macroscopic examination revealed a female fetus weighing 1500 g. The fetus exhibited a pronounced anterior coelosomy with externalized liver and intestines, thoracoschisis, and ectopiacordis. Limb anomalies included clubfoot, absence of the left upper limb, and syndactyly. No cerebral anomalies or cleft lip were observed. The placentawas connected to the fetus via placento-abdominal attachments **(Figure 3,4).**

Discussion:-
Limb Body Wall Complex (LBWC) is a rare and severe polymalformative syndrome, often underrecognized, making its true incidence difficult to ascertain. It is estimated to occur in 1 in 14,000 to 1 in 31,000 pregnancies according to various studies [26].

Pathophysiology
The exact etiology of LBWC remains poorly understood, and no specific teratogenic agent has been definitively implicated. Threeprimarytheories have been proposed:

1. Amnion Rupture Theory (Exogenous Theory):
Introduced by Torpin et al. in 1965 [7] and supported by later studies [8,9], this theory attributes LBWC to a primary rupture of the amnion, leading to the formation of amniotic bands. These bands inducefetal lesions through mechanical trauma.

2. Vascular Theory (Endogenous Theory):
Proposed by Van Allen et al. [1], this theory suggests that embryonic vascular disruption between 4 and 6 weeks of gestation causes hemorrhagic necrosis, anoxia, and tissue loss, resulting in impaired embryonic development and associated malformations.

3. Germinal Disc Defect Theory:
Initially proposed by Streeter in 1930 [10] and expanded by Hartwig et al. [11,12], this theory implicates a primary defect in the germinal disc, which disrupts embryonic folding and the development of the embryonic pedicle. This abnormality, occurring around day 32 of gestation, leads to abdominal wall closure defects, persistence of the extraembryonic coelom, and anomalies in the umbilical cord, placenta, and limbs [5,13].

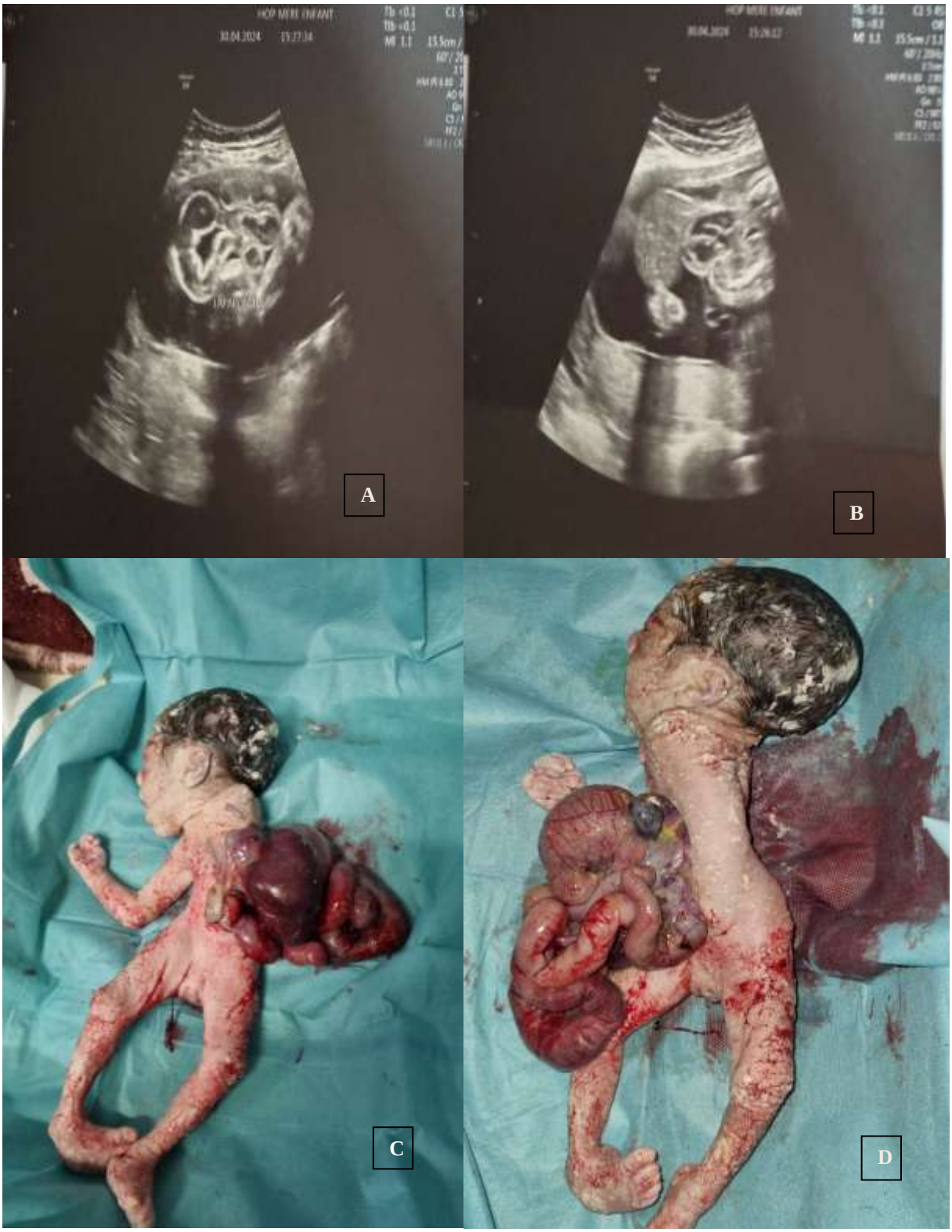

Figure 3:- (A-B) Longitudinal USG view showing the herniated abdominal contents**(**liver, bowel,ectopiacardis**),**
Figure 4:- (C) Post delivery image showing large defect in the anterior abdominal wall defect with liver and gut contents herniating through it,
(D) ectopiacardis, bilateral clubfeet deformity, ectrodactylie, left upper amelia.

Diagnostic Criteria

The diagnostic criteria for LBWC have been widely debated. Van Allen et al. suggested a diagnosis when at least two of the following anomalies are present [1]:

1. Exencephaly or encephalocele with facial clefts,
2. Thoracic and/or abdominal coelosomy,
3. Limb anomalies.

Building on these criteria, Russo et al. [5] and Cusi et al. [6] proposed two phenotypes based on placental attachment:

- **Cranial Placental Attachment Phenotype:**

Associated with neural tube defects in the cranial region, complex facial clefts, anterior coelosomy, and occasional amniotic bands. Limb anomalies, whenpresent, typically affect the upper limbs.

- **Abdominal Placental Attachment Phenotype:**

Characterized by complex urogenital anomalies, frequent spinal abnormalities, and limb anomalies primarily affecting the lower limbs [18].

Case Insights

Our first case is notable for the coexistence of cranial and abdominal placental attachments, a combination not previously described to our knowledge. This unusual presentation highlights the potential variability in LBWC phenotypes and raises questions about overlapping etiopathogenic mechanisms.

The second case exhibited abdominal placental attachment with limb anomalies, consistent with the abdominal phenotype described in the literature. These findings emphasize the importance of detailed phenotypic characterization in understanding LBWC and its underlying mechanisms

Diagnostic and Prognostic Considerations

The diagnosis of Limb Body Wall Complex (LBWC) can be supported by measuring maternal serum alpha-fetoprotein levels and conducting prenatal ultrasound, which may detect the condition as early as the first trimester [2]. Increased nuchal translucency during the first trimester has been recognized as a potential marker for this anomaly [23].

Cai-Hong Ye et al., in a study spanning March 2015 to October 2022 involving 35,486 pregnant women who underwent nuchal translucency screening between 11 and 13+6 weeks of gestation, identified and confirmed 18 cases of LBWC following medical termination [24,25]. The most commonly observed anomalies in these cases included coelosomy, limb malformations, placental attachment to the fetus, and spinal abnormalities [14,15].

Necropsy remains the gold standard for confirming the diagnosis, as it allows for a thorough evaluation of anatomical anomalies. Additionally, karyotyping is typically normal in LBWC cases, further distinguishing it from chromosomal abnormalities.

Early prenatal diagnosis is essential for differentiating LBWC from conditions such as isolated gastroschisis or omphalocele, which carry significantly better prognoses. Unlike these anomalies, LBWC has an extremely poor prognosis, with survival beyond birth being exceptionally rare. Only two cases of prolonged postnatal survival have been reported, both involving severe physical disabilities [11,12].

Given the severity and uniformly poor outcomes associated with LBWC, early diagnosis followed by medical termination of pregnancy is the preferred management strategy [3].

Conclusion:-

Limb Body Wall Complex (LBWC) is a fatal polymalformativefetal syndrome that requires heightened awareness among obstetricians and sonographers to ensure timely prenatal diagnosis and appropriate management. Early detection through ultrasound, supplemented by magnetic resonance imaging when necessary, enables accurate identification of this condition and facilitates informed decision-making regarding pregnancy termination [16].

Given its uniformly poor prognosis, early diagnosis is crucial, as medical termination of pregnancy is widely recommended in cases of LBWC. In select cases, a precise prenatal diagnosis may obviate the need for postnatal autopsy.

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