

# Prenatal diagnosis of a particular limb body wall complex\*

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## ABSTRACT

**Limb Body Wall Complex (LBWC) is a rare heterogenic group of fetal malformations including major defects of the ventral body (thoracoabdominal) wall associated with other anomalies including those of the limbs that may range from amelia to mild positional deformations, unusual craniofacial malformations, and a variety of visceral abnormalities that include the heart, lungs, genitourinary system. Two phenotypes are described according to the fetoplacental attachment. We report a particular case and iconography of LBWC associated with a giant sacral meningocele never described before. The patient was 26 years old, primigravida, with no illnesses or recent infections. She had no history of prenatal exposure to teratogenic agents, nor any family history of congenital malformations. She was referred for polymalformatif syndrome at 30 gestation weeks. Advanced sonography showed giant omphalocele with eviscerated liver, stomach and kidney, club feet, kyphoscoliosis and giant sacral fluid cyst. A 2100 g fetus was delivered, postnatal examination confirmed LBWC with abdominoplacental attachment and the particularity of the presence of giant sacral meningocele, two bilateral hemigenital tubercles and anal atresia.**

## KEYWORDS

**Fetal Malformation; Laparoscisis; Feto-Placental Attachment; Kyphoscoliosis**

## 1. INTRODUCTION

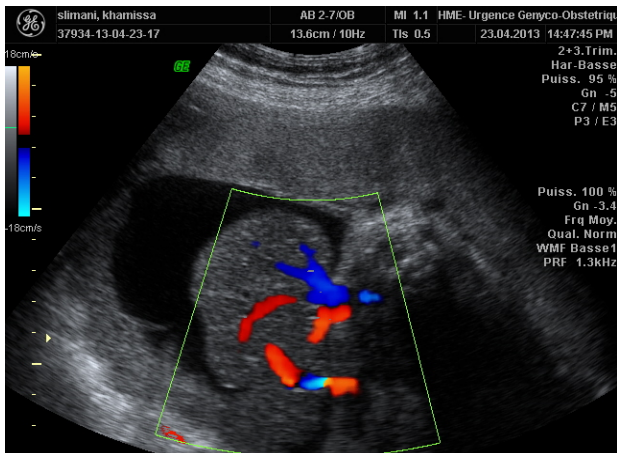
Limb-body wall complex (LBWC) is a rare, complicated, polymalformative fetal malformation syndrome including neural tube defects, body wall disruption and limb ab-

normalities [1]. The incidence at birth is about 0.32 per 100,000 births because the majority of affected fetuses undergo intrauterine deaths [2,3]. Pathogenesis is complicated and the possible pathogenetic mechanisms for LBWC include early amnion rupture, vascular disruption, and early embryonic maldevelopment. Generally the diagnosis is based on two of three of the following anomalies: Exencephaly or encephalocele with facial clefts, major defect of the ventral body wall (Thoraco and/or abdominoschisis) and Limbs anomalies that may range from amelia to mild positional deformations. Two phenotypes have been described, according to “placentocranial” or “placento-abdominal” adhesions [1,2]. Early ultrasonographic examination (12 weeks of amenorrhea) can make antenatal diagnosis [4]. Few cases are described, we present with iconography a case of LBWC with an interesting feature noted, consisting on the presence of a huge sacral meningocele never described before in the literature.

## 2. CASE REPORT

26 years old, primigravida, with no illnesses or recent infections. She had no history of prenatal exposure to teratogenic agents, nor any family history of congenital malformations. She was referred for polymalformatif syndrome at 30 gestation weeks. Advanced sonography showed giant omphalocele with eviscerated liver (**Figure 1**), stomach and kidney (**Figure 2**), club feet, kyphoscoliosis (**Figure 3**) and giant sacral fluid cyst. LBWC was suspected but the sacral cyst was an atypical association. A 2100g still birth fetus was delivered prematurely and spontaneously, will decision was not yet taken. Postnatal examination confirmed LBWC with abdominoplacental and amniotic attachment and the particularity was the presence of giant sacral meningocele, two bilateral hemigenital tubercles and anal arteria. Unfortunately placenta was attached during delivery but amniotic membranes was still attached to abdomen (**Figures 4 (a)-(c)**).

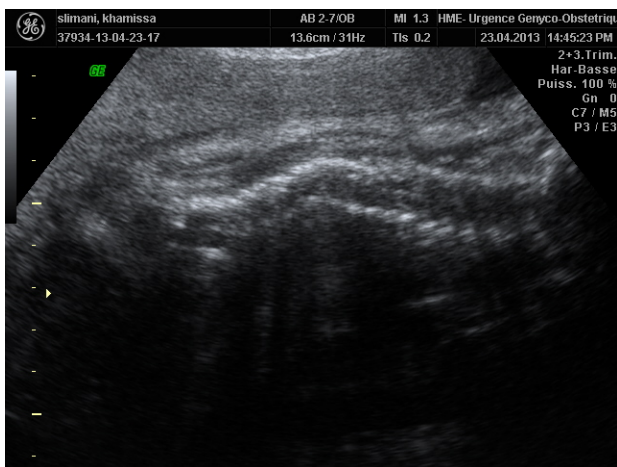
\*The case report was approved by gynecology obstetrics department ethics committee.



**Figure 1.** Colour flow imaging scans of the fetus showing a large ill defined abdominal wall defect through which the liver herniated into the extra embryonic coelom.



**Figure 2.** Ultrasound aspect with kidney protrusion through anterior abdominal defect.



**Figure 3.** Ultrasound aspect of the fetal kyphoscoliosis.

Feetopathology and caryotype was not done because inability in our centre.



(a)



(b)



(c)

**Figure 4.** General appearance after delivery.

### 3. DISCUSSION

The limb body wall complex is also known as the body-stalk syndrome [1]. It is a rare entity characterized by severe malformations. Most fetuses are aborted, either spontaneously or by medical means [5]. Most of them are stillborn. Postnatal survival for a significant duration is extremely rare, but at least one such case is reported; the child has severe physical handicaps [5]. This congenital anomaly has no sex or familial predilection or known recurrence risk. Caryotype study has been reported to be normal in all cases of LBWC. Van Allen [6,7], who described the syndrome for the first time in 1987 mentioned 3 essential features: 1) Exencephaly/encephalocele with facial clefts, 2) thoraco andabdominoschisis, 3) limb defect. In general, the diagnosis is based on two of the above features. Different authors described the two different phenotypes, according to the placental attachment: placento-cranial and placento-caudal types.

1) The placento-cranial attachment, showing craniofacial defects and amniotic bands and/or adhesion; it seems to be related to early vascular disruption.

2) The placento-abdominal attachment that presents urogenital anomalies, anal atresia, short umbilical cord, abdominal placental attachment and persistence of anextraembryonic celom. It seems to be due to an intrinsic embryonic maldevelopment.

Also, Torpin and all have suggested that LBWC results from amnion rupture caused by vascular or mechanical compression occurring between third and fifth weeks [7,8]. In our case report, the pattern of presentation resembled the "placento-abdominal" adhesion phenotype. Serum alpha-fetoprotein measurement and ultrasonographic examination are the key of prenatal diagnosis [9]. The ultrasound milestone of LBWC consists of thoraco-abdominal defect, spinal and cord abnormalities, positional limb deformities and abnormalities of umbilical cord and membranes. All these groups of defects were present in our case. The severity of the defects and a distorted appearance of the fetus make recognition of the fetal parts difficult in almost all cases of LBWC.

LBWC should be differentiated from common abdominal wall defects such as gastroschisis, omphalocele and uncommon entities like ectopic cordis, amniotic band syndrome, cloacal dystrophy and urachal cyst. There are also some specific signs for LBWC: the presence of membranes covering the contents of the herniated sac, any associated bowel abnormalities, the presence or absence of urinary bladder, scoliosis and limb defects [6, 9].

In our case, we report a huge sacral meningocele undescribed before in the literature.

### 4. CONCLUSION

LBWC is a rare polymalformative syndrome possible to detection with antenatal ultrasound scan. The syndrome is invariably fatal, but the couple must be reassured that there is no risk for recurrence. The diagnosis can be established early, thanks to the fascinating progress of sonographic technology and followed by medical termination of the pregnancy.

### CONFLICT OF INTEREST

There is no conflict of interest.

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