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## Case Report

# A rare case of limb body wall complex<sup>☆,☆☆</sup>

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### ABSTRACT

Limb body wall complex (LBWC) is a complex and rare poly-malformative syndrome. We report a case of this syndrome diagnosed antenatally in a 37-year-old primiparous woman, by a first obstetrical ultrasound performed at 22 weeks of amenorrhea and one day. After termination of pregnancy, macroscopic examination revealed a male newborn with all the diagnostic criteria of LBWC.

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## Introduction

Limb body wall complex (LBWC) is a rare specific poly-malformative syndrome, little known by obstetricians and sonographers until 1987. It was described more precisely by VAN Allen et al. [1]. It associates severe anterior parietal, visceral, encephalic spinal and limb malformations [2]. Antenatal diagnosis can be made by antenatal ultrasound, provided that it is recognized. For this purpose, we report a case of antenatal diagnosis of this syndrome made at 22 weeks of amenorrhea and one day, in a primiparous woman, specifying the specific features of our observation and we try to recall the diagnostic criteria, the etio-pathogenic mechanisms and the prognosis of this syndrome, which is not as rare as previously believed.

## Case Report

This is a patient with no particular medical or surgical history. She has been married for 2 years, with no notion of consanguinity or family fetal malformation. Primigravida, unattended pregnancy, estimated at 5 months, with no notion of specific medication, referred for poly-malformative syndrome. The clinical examination revealed a uterine height corresponding to the gestational age with positive BCF. Obstetrical ultrasound revealed an evolving mono-fetal pregnancy, amniotic fluid in normal quantity, a large encephalocele with unilateral ocular agenesis (Fig. 1), associated with a complex cleft lip, a large upper coelosomia, heart, liver, (Fig. 2) without individualization of diaphragm, a femur length measured at 31.2 mm corresponding to 19 SA+4d with associated suspicion of amniotic band disease and the presence of a single artery at the level of the umbilical cord (Fig. 3). The most likely

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**Fig. 1 – Coronal section showing ocular agenesis (→).**



**Fig. 2 – Sagittal section showing an anterior coelosomy (→).**

diagnosis was LBWC. A medical termination of pregnancy was performed after discussion with the parents. Macroscopic examination at birth showed a male fetus weighing 380 g, with an encephalocele with a large asymmetrical cleft lip, a very marked anterior coelosomy (heart, liver, and intestine exteriorized) and a narrow thorax. A narrow thorax (Fig. 4). Unfortunately, the anatomopathology examination in search of other visceral malformations could not be performed because we do not yet have a fetopathology unit. However, all the ultrasound

findings and the gross examination allowed us to make the diagnosis.

### Discussion

LBWC is a complex poly-malformative syndrome, insofar as it is not always recognized as such, its incidence is difficult



**Fig.3 – Single umbilical artery objectified by color Doppler.**



**Fig. 4 – Photo of the newborn with Limb Body Wall Complex with poly-malformative syndrome: Encephalocele (→), Complex cleft (☆), Anterior coelosomy (⇔) Weight : 380g.**

to estimate: located between 0.21 and 0.31 per 10,000 births according to the authors [3,4]. About 250 cases have been reported in the literature [3,5,6].

Its physio-pathogenesis is debated and poorly known. Most studies do not implicate any teratogenic agent. There are three opposing theories. The theory of hyper-early rupture of the amnion or "exogenous" theory was introduced by Torpin et al. in 1965 [7] and taken up by several authors [8,9]. It implies

a primary interruption of the amnion inducing the formation of amniotic bridges, which would be by a traumatic phenomenon, themselves responsible for the fetal lesions. This is the most probable theory for our case. Van Allen et al [1], propose a second hypothesis: the vascular or "endogenous" theory by infarction of embryonic vessels between 4 and 6 weeks of gestation leading to tissue losses, an interruption in the development of embryonic tissue due to hemorrhagic necro-

sis and anoxia, with deformations. Streeter in 1930 evoked a primitive defect of the germinal disc [10], a concept developed in particular by Hartwig et al. [11,12] under the term of the embryonic closure anomaly theory. They incriminate an anomaly in the development of the embryonic pedicle with disruption of the caudal and lateral folds of the embryonic disc from 32 days of gestation. This leads to an anomaly in the closure of the abdominal wall, the persistence of the extra-embryonic coelom; anomalies of the cord, placenta and limbs [5,13]. However, none of these different theories seems to explain all the syndromes on its own.

The diagnostic criteria for LBWC are much debated in the literature. Initially, Van Allen et al proposed that LBWC should be diagnosed when at least two of the following three types of anomalies were present [1]: exencephaly or encephalocele with facial clefts, thoracic and/or abdominal coelosomy and limb anomaly.

More recently, Russo et al. [5] and Cusi et al. [6] have considered two different phenotypes, depending on the type of placental attachment. Phenotype with placental-cranial attachment which associates: a neural tube closure defect in the cephalic territory, always associated with one or more complex facial clefts, anterior coelosomy, and inconstant amniotic flanges. Limb anomalies are facultative and mainly affect the upper limbs.

Phenotype with placento-abdominal attachment characterized mainly by complex urogenital anomalies, frequent spinal anomalies. The anomalies are optional and affect mainly the lower limbs.

Antenatal diagnosis by ultrasound is possible from the end of the first trimester, at best vaginally, the earliest antenatal diagnosis in the literature is made at 12 SA [2]. The most frequently reported anomalies are coelosomia, limb involvement, placental attachment to the fetus and spinal anomalies [14–16]. The differential diagnoses to be considered would be numerous if each anomaly was considered in isolation, diagnostic difficulties may be encountered in case of severe oligohydramnios as well as in the case of association of an omphalocele or laparoxisis with other malformations such as neural dysgraphia or craniofacial defects [13,17]. In our observation, our diagnosis was mainly based on the association of a complex poly-malformative syndrome with the suspicion of amniotic band disease.

The prognosis of LBWC is fatal, the malformations being unfortunately beyond therapeutic resources, a termination of pregnancy seems acceptable in all cases. Given the absence of chromosomal abnormalities, a karyotype seems unnecessary. The risk of recurrence of the condition was considered nil until 2002 when Leurh et al. reported 2 cases of familial recurrence that could lead to discussion of a probable genetic origin [18].

## Conclusion

The limb body wall complex syndrome deserves to be better known by obstetricians and ultrasonographers because it allows both to make an antenatal diagnosis and to discuss an early termination of pregnancy, but above all to reassure cou-

ples facing such a poly-malformative syndrome because the risk of recurrence is almost zero.

## Patient consent

The patient gave consent for publication.

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