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Case Report

Limb Body Wall Complex: A Case Report

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Abstract

Limb-body wall complex (LBWC) is a rare, congenital defect defined by lateral body-wall defects, limb reduction abnormalities and/or craniofacial defects. The developmental pathogenesis as well as the etiology of LBW complex is controversial and has no sex or familial predilection. The poor prognosis of LBWC necessitates an early antenatal diagnosis. Serum alpha-fetoprotein measurement and ultrasonographic examination is the key to prenatal diagnosis**.** Prenatally, the abnormal fetoplacental attachment can be detected ultrasonographically by the end of the first gestational trimester. Postnatal, the examination of placenta, umbilical cord and membranes is crucial in confirming the diagnosis of LBWC. The present case is associated with amniotic adhesive bands, thoraco-abdominoschisis, minor encephalocele, and belongs to the category of LBWC with craniofacial defects.

Keywords: Abdominal wall defect - Limb-body wall complex - Placenta - Umbilical cord.

INTRODUCTION

Limb body wall complex (LBWC) also known as “body stalk syndrome” is a rare complicated sporadic polymalformative fetal malformation syndrome, refers to a variable group of congenital defects characterized by a wide spectrum of severe anomalies in the body wall, with essential features of: Exencephaly and/or encephalocele with facial clefts, Thoraco and/or abdominoschisis, and Limb defects [1].

The incidence at birth is about 0.32 per 100,000 births because the majority of affected fetuses undergo intrauterine deaths [2].

Two phenotypes have been described, the “placento-cranial” and “placento-abdominal” adhesion phenotypes. The developmental pathogenesis as well as the etiology of LBW complex is controversial [3] and has no sex or familial predilection.

Three mechanisms have been proposed: early amnion rupture [4-8] embryonic circulatory failure leading to vascular disruption [9, 10] and embryonic dysgenesis due to malfunction of ectodermal placodes [11-14] or to localized disturbance of basic embryonic organization.

LBWC is almost uniformly lethal and associated with a very poor outcome because of, among other factors, pulmonary hypoplasia [15-18]. Which is why the early antenatal diagnosis is necessary. Serum alpha-fetoprotein measurement and ultrasonographic examination is the key to prenatal diagnosis.

CASE REPORT

We report the case of patient S, A 30 year-old gravida 4, para 3 with no particular medical, obstetrical , or surgical history and with no notion of consanguinity or family fetal malformation.

She did not have a history of recent infections or exposure to teratogen agents (excepted the notion of taking food made of fenugreek), and there was no family history of congenital malformations. She had not undergone assisted reproductive technology for this pregnancy.

The pregnancy was never followed until the 36 Th week of gestation, when the membranes ruptured spontaneously.

The patient was transferred to our department to get a more detailed evaluation in the presence of a suspected diagnosis of disease after ultrasonographic examination suggested a severe abdominal wall defect.

The clinical examination revealed a uterine height corresponding to the gestational age, a premature rupture of the membranes with a cervix dilated to 2 cm, and a positive and regular cardiac activity.

Ultrasound examination shows a live male fetus of 36 weeks old according to ultrasound measurements, with a minor, Anamnios, abdomino-placental attachment, and thoraco-abdominal wall defects with a cardiac ectopy but with no limb deficiency (Fig 1).

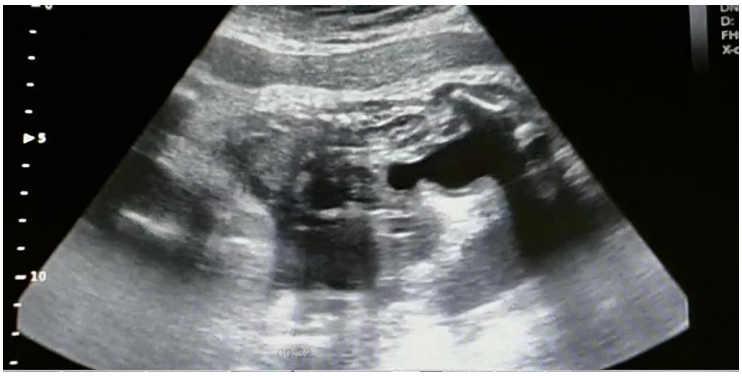

Fig 1: Ultrasound appearance of large anterior wall defects in a fetus at 36 SA with a Limb Body Wall Complex, at the Souissi maternity hospital in Rabat

An emergency cesarean section was indicated in the presence of fetal bradycardia that allowed the extraction of a 2100-g male fetus with abdonio-placental attachment, craniofacial deformity with multiple amniotic bands around the skull, scoliosis, and a large thoraco-abdominoschisis with cardiac ectopy, eviscerated liver, stomach and bowel, with a normal ano-genital region (Fig 2).

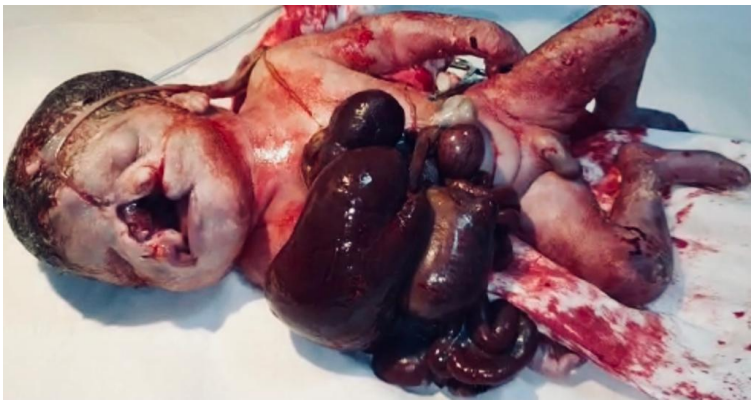

Fig 2: External appearance of Newborn with Limb Body Wall Complex with poly-malformative syndrome: encephalocele with amniotic bands, complex cleft, large anterior wall defect, born at the Souissi maternity hospital in Rabat

The umbilical cord was short, straight, incompletely covered by amnion and adherent to the placental membranes as well as the eviscerated mass.

A diagnosis of limb body wall complex (LBWC) with craniofacial defects was made. Unfortunately the newborn died shortly after birth due to the severity of the malformations and defects.

DISCUSSION

LBWC was described for the first time in 1987, it is also known by the "Body stalk anomaly" "Congenital absence of umbilical cord" and "cyllosomus and Pleurosomus" [19, 20] . It describes a heterogenic group of fetal malformations, including lateral body-wall defects and limb reduction anomalies [9, 21].

The first phenotype shows craniofacial defects and amniotic bands and/or adhesion, while the second is without craniofacial defects and presents urogenital anomalies, anal atresia, and abdominal placental attachment, together with a persistence of the extra-embryonic celom [22].

In 2007 Sahinoglu *et al*., [23] proposed a new classification: Type I (Fetus has craniofacial defect and intact thoracoabdominal wall, often normal placenta and umbilical cord but rarely attached to the malformed cranial structures), Type II (Fetus has supraumbilical, usually laterally located, often left side, large thoracoabdominal wall defect).

The internal organ malformation present in 95% cases was cardiac defects, absent diaphragm, bowel atresia, renal agenesis and renal dysplasia. Body defects was a central feature of LBWC.

In our case report, the pattern of presentation resembled the "placento-abdominal" adhesion phenotype.

The possible pathogenetic mechanisms for LBWC include early amnion rupture [25], vascular disruption [24, 21], and early embryonic maldevelopment [26, 12]. LBWC with CF defects is caused by an early vascular disruption, whereas LBWC without CF defects is related to defective lateral and caudal folding of the embryonic disk [22]. Recent reports have suggested that assisted reproductive technology increased the risk of congenital malformations, compared with natural conceiving infants. Pre-ovulatory administration of clomiphene citrate to mice has been shown to impair uterine functions and cause fetal growth retardation and neural tube defects in post-implantation embryos [28]. Ovarian stimulation may also increase the risk of imprinting disorders [29, 30].

Prenatal ultrasound examination can detect this anomaly as early as first trimester (usually by the end of first trimester) [19]. The principal findings are the thoracoabdominal defect, limb anomalies, spinal and cord abnormalities [22]. 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. The diagnosis of this condition can be also established by measuring maternal serum alpha fetoprotein level or fetography that provides a clear outline of the fetal soft tissue and the three-dimensional structure of the fetus and membrane, making it a very useful technique for the diagnosis of this pathology.

For the confirmation of the diagnosis, the autopsy remains the gold standard. Caryotyping is usually normal. Early diagnosis followed by medical termination is the preferred treatment for this anomaly [19].

CONCLUSION

LBWC is a rare congenital anomaly with no sex predilection, it has a possible genetic cause, but the most accepted theory is "early embryonal dysplasia." It is important to differentiate them from other anterior abdominal wall defects.

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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