Early diagnosis of limb body wall complex: a case report

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Summary

Limb body wall complex (LBWC) is a rare polymalformative syndrome. The majority of cases presented in the literature have been diagnosed in the second or third trimester of pregnancy. The authors present a case of LBWC diagnosed in the first trimester of pregnancy. The combination of two- and three-dimensional ultrasounds proved to be useful for establishing the diagnosis and differentiating from other abdominal wall defects. The diagnosis was followed by therapeutic abortion at 14 weeks' gestation, and the pathological report confirmed the diagnosis.

Key words: Limb body wall complex; Umbilical cord syndrome; Body stalk anomaly; Two-dimensional ultrasound; Three-dimensional ultrasound.

Introduction

The limb body wall complex (LBWC), a rare polymalformative syndrome, first described by Van Allen is diagnosed based on the presence of at least two of the three following characteristics: 1) exencephaly/encepalocele and facial clefts; 2) thoraco-and/or abdominoschisis, and 3) limb defects (1,2). The authors present a case of LBWC diagnosed early during pregnancy using two-dimensional (2D) three-dimensional (3D) ultrasonography.

Case Report

A 24-year-old woman, gravida 1, para 0, and abortus 0 presented for ultrasound for nuchal translucency scanning at 12 weeks and five days of amenorrhea. The woman had a normal history, and neither her family nor the baby's father's family had a history of genetic anomalies or malformations. The ultrasound revealed a large defect in the anterior abdominal wall (Figure 1). The fetal head showed exencephaly with evidence of scoliosis. The umbilical cord was very short. Both the spinal and abdominal wall defects were clearly visible with 3D ultrasound (Figure 2).

Based on the ultrasound features, a diagnosis of LBWC was formulated. The patient was informed of the poor prognosis and counseled about the condition. The 3D ultrasound images proved useful in helping the patient understand the syndrome. Because of the gestational age, after counseling, the patient decided to have therapeutic abortion at 14 weeks' gestation.

The diagnosis was confirmed by anatomopathologic examination (Figures 3, 4). The liver and intestines were outside the body. Fetal head showed exencephaly and cleft lip. The superior right arm was shortened and had an aberrant site of implantation. There was no right forearm and the right hand had only three fingers. Usually limb anomalies described in the literature are more frequent in the inferior arms. The umbilical cord was short, straight, incompletely covered by amnion and adherent to the placental membranes, as well as eviscerated mass. After the abortion the parents were counseled that there was no risk of recurrence for the subsequent pregnancy. The woman gave birth after one year to a healthy newborn (female, 3,200 g, at 38 weeks gestation).

Discussion

LBWC, a rare polymalformative fetal syndrome anomaly of the anterior wall with a very poor prognosis, has an incidence of 0.21 to 0.31 per 10,000 live births (3). Diagnosis is based on the presence of at least two of three essential features: exencephaly/encepalocele and facial clefts, thoraco-and/or abdominoschisis, and limb defects (1,2). A constant finding is coelosomia, which can be variably associated with encephalic, vertebral, visceral or limb anomalies. The anomaly has also been referred to in the literature as a short umbilical cord syndrome or body stalk anomaly.

Some authors assert that LBWC simply represents a severe form of amniotic band syndrome. Indeed, in almost 40% of cases, amniotic bands are present and the defect of the limbs could appear to be secondary to the amniotic bands. The pathogenesis of LBWC is unclear, and, different pathogenic mechanisms have been suggested. Several theories have attempted to explain the syndrome: germ disc defect with early embryonic maldevelopment (4,5), early amnion rupture theory (6,7), vascular disruption theory (8,9), and the disturbance of the embryonic folding process (4). Hunter *et al.* claimed a new theory in which association of malformations originates as early as the embryonic disc stage and that some of the associated anomalies are secondary complications of the primary disturbance in embryogenesis (10). LBWC will always appear as a combination of a large abdominal wall defect with protrusion of the viscera, a severe spinal scoliosis and a continuous juxtaposition of the fetus to the placenta. The eviscerated organs form a complex mass entangled with membranes. The defect comprises most often both the ab-

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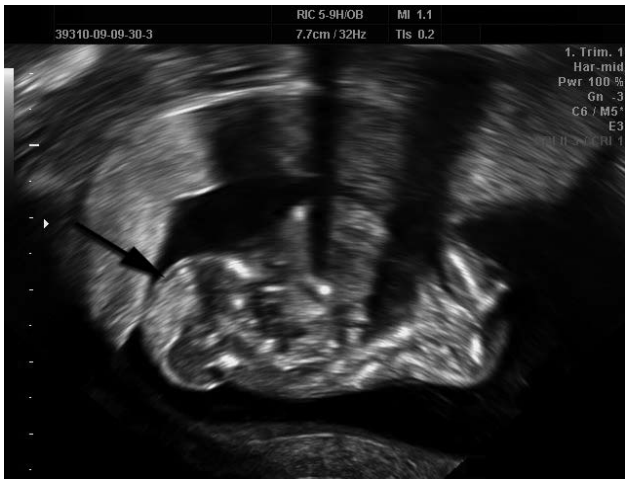

Figure 1. — 2D ultrasound of the fetus showing a large defect of the abdominal anterior wall. The cephalic extremity is not well-defined and has a abnormal shape and spine kyphosis is present.

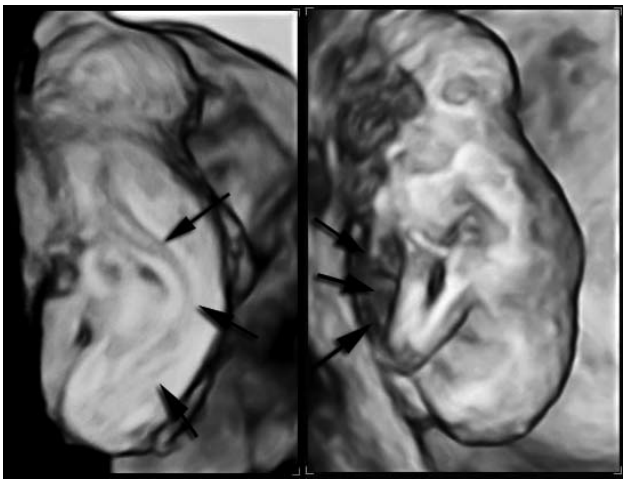

Figure 2. — 3D ultrasound showing severe scoliosis of the spine with a large defect of the anterior abdominal wall.

Figure 3. — The fetus after abortion (ventral view) with exencephaly, large abdominal defect, anomalous implantation of the right arm, missing forearm, oligodactyly, and short umbilical cord.

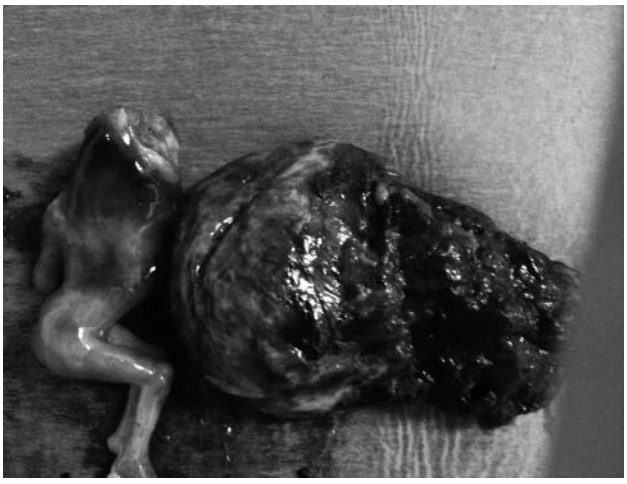

Figure 4. — The fetus after abortion (dorsal view) with a marked spine scoliosis.

domen and the thorax or rarely could be limited only to the abdomen or thorax. Associated malformations include central nervous system lesions, facial abnormalities, cardiac malformations, urogenital anomalies, limb defects, amniotic bands, and placental abnormalities.

The limb defect can manifest as clubfoot, oligodactyly, arthrogryposis, and absent limbs or digits. Severe scoliosis develops as a consequence of the irregular attachment of the fetus to the placenta. Russo *et al.* identified two distinct LBWC phenotypes: placento-cranial and placento-abdominal types (11). The placento-cranial type is characterized by craniofacial defects, facial clefts, and amniotic adhesions. The placento-abdominal type has no cranial defects, rather, more frequently has been associated with coelosomia lumbosacral meningomyelocele and kyphoscoliosis, as well as urogenital anomalies. The present case has the characteristic of the former type of LBWC. Usually, LBWC is diagnosed during the second trimester of pregnancy by ultrasound. Although the syndrome can also be diagnosed in the first trimester of pregnancy, very few cases of diagnosis in early gestation have been reported (12-14). Nonetheless, the diagnosis cannot be formulated before ten weeks' gestation because of the physiological herniation characteristic of this period. In the present case, the diagnosis was established at the end of the first trimester of pregnancy. The presentation of this case clearly showed the importance and the benefit of a correct prenatal diagnosis. Because the ultrasound examination was correctly performed, this condition was diagnosed early and a therapeutic abortion was performed.

It is important to distinguish LBWC from other anterior wall defects to determine the prognosis and management options (15-17). The site of the defect should be considered when differentiating LBWC from other abdominal wall anomalies. In ectopia cordis, the defect is situated at the abdominal wall and anterior aspect of the thorax, but also involves the heart, while cloacal extrophy involves the lower abdominal wall (14). Also, it is important to discern LBWC from isolated gastroschisis, which has a much better prognosis (15).

The prognosis for LBWC is very poor and because it is considered incompatible with life, pregnancy should be terminated after a correct sonographic diagnosis. It is also important to explain to the families affected that there is no risk of recurrence of this anomaly.

Conclusion

The present case demonstrated the importance of performing a thorough morphologic survey at the time of nuchal translucency screening, and the value of 3D sonography in the delineation of first-trimester anomalies. Also, 3D ultrasound is a valuable tool for counseling parents to provide a better understanding of the problems that could appear.

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