

Prenatal ultrasonographic diagnosis of limb body wall complex

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Abstract

In this report, a case of limb body wall complex (LBWC) diagnosed by ultrasonography is presented. Limb-body wall complex refers to a rare combination of disruptive and lethal abnormalities which start early in the gestational process. Abnormalities commonly associated with this disorder include cranio-facial abnormalities, scoliosis, ventral body wall defect (thoraco-abdominoschisis), limb deformations, short umbilical cord, and others. Other terms used to describe similar findings include short umbilical cord syndrome, body-stalk anomaly, and amniotic band syndrome. This complex should be distinguished from other body-wall defects including omphalocele and gastroschisis since the prognosis for limb-body wall complex is uniformly poor. The diagnosis of limb-body wall complex can be made by prenatal sonography.

Key words: limb body wall complex, abdominal wall defects, amniotic bands, amniotic band syndrome, ultrasonography.

Introduction

Limb body wall complex is rare, complicated, polymalformative fetal malformation syndrome with essential features of a) exencephaly/encephalocele with facial clefts b) thoraco-and/or abdominoschisis and c) limb defect. Generally, the diagnosis is based on two of three of above features.¹ It has been proposed that this disorder occurs from early amniotic rupture with direct mechanical pressure and amniotic strands producing the typical defects.² Besides, it has been reported that this complex occurs as a result of defect in germ disc causing abnormal formation in amniotic cavity, or this complex is seen as a result of teratogenic effect during early pregnancy. It was also pointed out that this complex occurs as a result of vascular abnormality during early embryonal period.³ This complex malformation has no sex or familial predilection and is invariably fatal. The poor prognosis of LBWC necessitates an early antenatal diagnosis. Maternal serum alpha-fetoprotein (MSAFP) measurement and ultrasonographic examination is the key to prenatal diagnosis. The ultrasonographic hallmark of the LBWC is characterized by thoraco-and /or abdominoschisis, neural-tube abnormalities, severe

scoliosis, positional deformities and abnormalities of fetal membranes.⁴ In this case report we highlight the ultrasonographic finding of LBWC diagnosed on antenatal scanning along with photographs of aborted fetus.

Case

A 23 year old primigravida was referred at 32 weeks of gestation for antenatal ultrasonographic scanning. She booked case with history of non-sanguinous marriage (not on any medications).

Ultrasound scan revealed a live single fetus with biparietal diameter corresponds to 29 weeks. The scan also revealed a large ill-defined abdominal wall defect through which the abdominal contents herniated into extra abdominal embryonic coelom, so abdominal diameter was disproportionately reduced (Fig1). There was also the evidence of extremity abnormalities. No anomalies were seen at the fetal eyes, facial profile, palate, lips, neck, spine, fetal heart, lung and urogenital structures. Placenta was fundal and right lateral in position with two arteries and one vein in umbilical cord. Since there was gastroschisis with extremity

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Figure 1. Ultrasonogram showing floating bowel loops into amniotic fluid due to defect in anterior abdominal wall of the fetus.



Figure 2. Herniation of abdominal contents including liver through a large defect.



Figure 3. Photograph showing hypoplastic and underdeveloped right arm with single finger and clubfoot with syndactyly.

abnormalities in ultrasound, provisional diagnosis of LBWC was made.

The patient was informed of the poor prognosis and after counseling, the patient elected to terminate the pregnancy. The pregnancy was terminated with the birth of a female fetus along with complete placenta and membranes.

The diagnosis of LBWC was confirmed after delivery. The detail examination of the fetus revealed herniation of abdominal contents through a large defect (Fig 2). The eviscerated organ formed a complex, bizarre appearing mass. An extracorporeal liver was present. The extremity abnormalities were hypoplastic and underdeveloped right upper limb with single finger (Fig 3). Both lower limbs showed clubfoot with syndactyly (Fig 3). There was also evidence of scoliosis. The umbilical cord was short; however cut section showed two arteries and one vein.

After delivery she was informed that there was no familial predilection or no known recurrence risk. She was counseled to report to hospital for regular screening in her future pregnancies.

Comments

LBWC is a rare complicated, polymalformative fetal malformation syndrome. In our case, there were anomalies in extremities and abdominal wall defects, especially gastroschisis have called attention. LBWC was diagnosed on the basis of abdominal wall and limb defects. The pathogenesis of LBWC is unclear and uncertain. Three pathogenetic mechanisms have been proposed namely, the early amnion rupture theory, vascular disruption theory and embryonic dysgenesis. Some consider that LBWC simply represents a severe form of amniotic band syndrome. This is reinforced by the presence of amniotic bands in nearly 40% cases of LBWC.⁵ More recently it is suggested that the presence of amniotic bands is a consequence of early vascular disruption, but is not the primary abnormality itself.³

In a study of a series of fetuses with LBWC the major structural defects included limb defects (95%), marked scoliosis (77%), internal organ malformation (95%), craniofacial defects (56%) and limb defects including club foot (32%). Body defect was a central feature of LBWC and it was found in 96% of cases. The involvement of both abdomen and thorax was a more common feature as compared to involvement of either abdomen or thorax solely. The abdominal and thoracic contents herniate through a large defect into the extra embryonic coelom. Typically, the eviscerated organs form a complex, bizarre appearing mass entangled with membranes⁵. In our case body defect was limited to abdomen and limb defects and scoliosis were also present.

The poor prognosis of LBWC necessitates an early antenatal diagnosis. Ultrasonographic detection of abdominoschisis, scoliosis, abnormalities of the extremities, a single umbilical artery, short umbilical cord and extremely elevated level of MSAFP is the key to early diagnosis. Ultrasonographically, the principal findings are the thoracoabdominal defect, limb anomalies, spinal and cord abnormalities.⁶ Almost all these groups of principal finding were present in our case.

Because LBWC is incompatible with life, it is important to diagnose the case prenatally and to differentiate it from anterior abdominal wall defects.⁷ LBWC should be differentiated from common abdominal wall defects such as gastroschisis, omphalocele and uncommon entities like ectopia cordis, amniotic band syndrome, cloacal dystrophy and urachal cyst. One of the principal parameters that assist this differentiation is the site. While ectopia cordis is typically located at anterior aspect of the thorax, gastroschisis and omphalocele are localized to the umbilical and paraumbilical area. Similarly cloacal dystrophy and urachal cyst are entities involving the lower abdominal wall. Other useful parameters that aid at deriving a specific diagnosis include the presence of membranes covering, the contents of herniated sac, any associated bowel abnormalities, the presence or absence of urinary bladder, scoliosis and limb defects, the presence or absence of other random defects such as facial cleft.⁵

Conclusion

Ultrasonographic detection of abdominoschisis and extremities abnormalities is important for prenatal diagnosis of LBWC. It is possible that LBWC might be confused with abdominal wall defects. Since LBWC

has a very poor prognosis, it is important to differentiate it from anterior abdominal wall defects.

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