LIMB BODY WALL COMPLEX (LBWC): A CASE REPORT OF INTRAUTERINE DIAGNOSIS BY ULTRASOUND IN SONGKHLA HOSPITAL

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ABSTRACT

A demonstrating case of intrauterine diagnosis of abdominal wall defect associated with multiple anomalies including extraabdominal wall mass contained fetal bowel, heart, and abdominal solid organ, unilateral dilatation of lateral ventricle, spinal and limb defect in Songkhla Hospital is reported. Discussion of the differences between omphalocele, gastroschisis and clinical features of limb body wall complex (LBWC) are also discussed.

INTRODUCTION

Limb body wall complex defines to a group of congenital defects having in common abdomino-or thoraco-schisis and multiple anomalies including craniofacial defect, body wall defect, spinal defect, limb defect and malformations of internal organs. Body walll defect can be recognized by detection of extrathoracic location of the heart (ectopia cordis) in case of thoracic wall defect or extra-abdominal mass if the defect involving the abdominal wall. Ectopia cordis can be diagnosed with ultrasound by visualizing the heart outside the thoracic cavity. The abdominal wall defect can result in either gastroschisis or omphalocele. The prenatal sonographic defferentiation of gastroschisis from omphalocele is sometimes unclear, eventhough the two entities are reported to be embryologically different. It is prognostic importance to establish a correct diagnosis because an isolated abdominal wall defect can be surgically corrected with excellent result while LBWC is usually fatal.

CASE REPORT

A 27-year-old primigravid woman was referred to perform ultrasound from a clinic. A 24-week fetus was detected that had a large extraabdominal mass measuring 8x10 cm containing fetal bowel and abdominal solid organ covering by a thin line of hyperechoic sac. The heart was also noted in cephalad portion of the mass (Fig. 1.), unilateral diltaion of lateral ventricle, unequal number of spinal ossification center in thoracic level and mild scoliosis, absence of normal fetal position. As multiple anomalies were found, the prognostic consideration was poor. Pregnancy termination was done by hypertonic normal saline induction with maternal approval. 1 day after induction, a 900 gram nonviable female fetus was delivered. Multiple anomalies included large head (episiotomy had to be done because of large head), large abdominal wall defect (totally absent abdominal wall since xyphoid) with fetal bowel, all abdominal organ and heart protruding outside covered by a thin layer of membrane. Other anomalies were short limbs, club foot and syndactyly. The mother was discharged 3 days later with approval and without complication.

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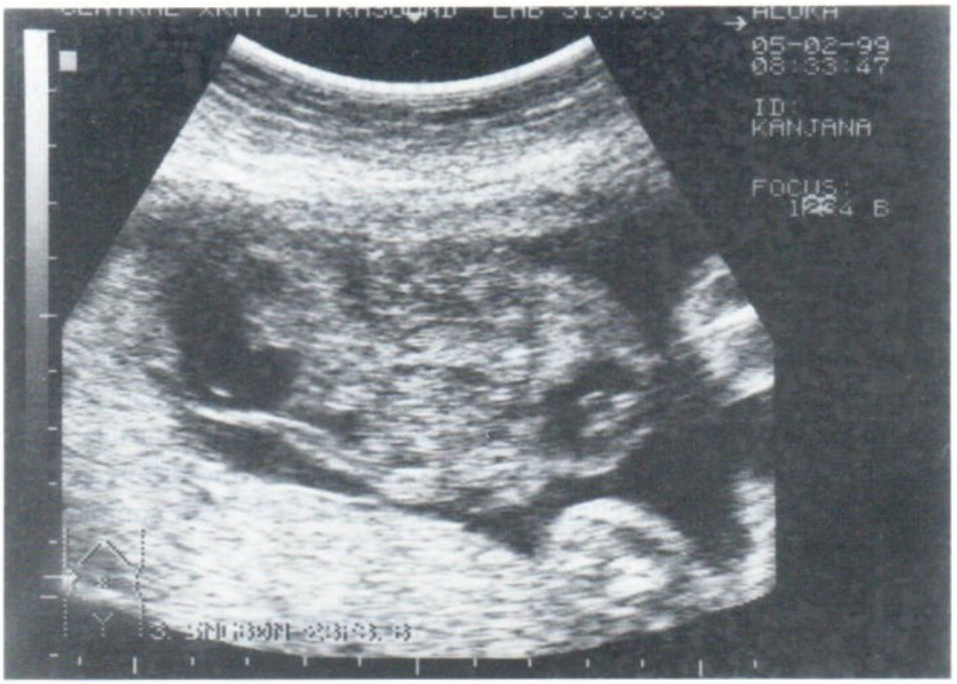

Fig. 1. Mixed echoic mass contained fetal bowel, abdominal solid organ and heart covering by a sac and appear contained rather than freely floated in amniotic fluid.

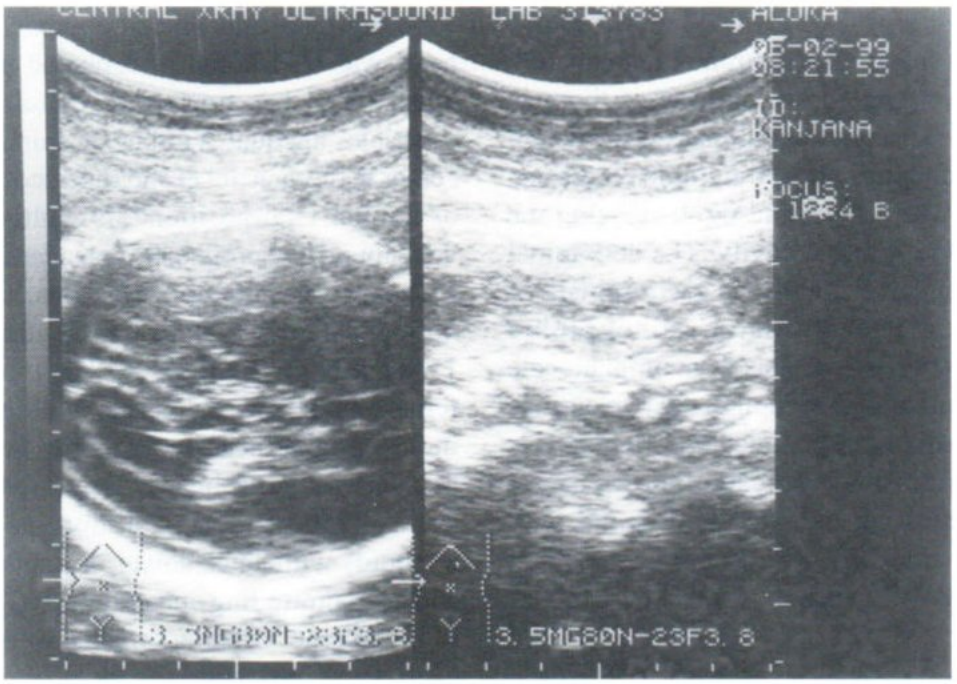

Fig. 2. (left) Unilateral dilatation of lateral ventricle. (right) unequal number of spinal ossification center and scoliosis.

DISCUSSION

Limb body wall complex defines to a group of congenital defects having in common abdomino-or thoraco-schisis and a complex anomalies. It is a complex malformation that has no sex or familial predilection or known recurrence risk.[2] The pathogenesis of the LBWC is uncertained. The primary pathological features include craniofacial defect (include exencephaly, encephalocele or facial cleft), a body wall defect involving the thorax, abdomen or both (64%), limb defects (95%) (including club foot (32%), absent limb or digits, arthrogryposis or web, polydactyly and syndactyly), malformation of internal organs (95%) (including cardiac defects (56%), absent diaphragm (74%), bowel atresia (22%), renal abnormalities (including agenesis, hydronephrosis or dysplasia (65%)) and spinal defects (including spinal dystrophic defects and/or scoliosis (77%)). Scoliosis is usually present in most cases of LBWC. Therefore the combination of ompha-locele and scoliosis should always suggest the diagnosis of LBWC. The prognosis of LBWC is invariably fatal.

On revision of embryogenesis, the anterior abdominal wall develops as the embryonal folds in both cranio-caudal and lateral directions. The cranial folds development at the 5th menstrual week. The heart and pericardium become to lie in the ventral surface and later incorporated into the chest when the lateral fold develops in the thoracic region. Parts of the yolk sac is also incorporated into the embryo as the midgut while ventral abdominal wall and lateral wall are forming. Coalescence of the body stalk with the yolk stalk forms the umbilical card at 7-8th menstrual week. Rapid expansion of the amniotic cavity obliterate the extra-embryonic coelom and forms the epithelial covering umbilical cord. About 8th menstrual week, rapid elongation of the midgut loop physiologically herniates into the base of umbilical cord and return to abdomen by 10-12th menstrual week with totally 270 degree counterclockwise rotating around the superior mesenteric artery. The final integrity of the abdominal wall depends on the fusion of these body folds together at the base of umbilical cord after the return of the intestines to the abdominal cavity. Failure of growth and fusion of cephalic fold result in either ectopia cordis or Pentalogy of Cantrell (cleft sternum, absence of diaphragmatic septum transversum, epigastrium omphalocele, ectopia cordis and a variaty of cardiovascular malformation).

The pathologic features of LBWC is the failure of fusion of the amnion and chorion. The amnion dose not cover the umbilical cord normally, but extends as a sheet from the margin of the cord and is continuous with both the body wall and the placenta.[3] Smith and others have suggested that LBWC results from rupture of the amnion caused by vascular disruption or mechanical compression between the third and fifth week.[5] Craven-CM and et al proposed that the pathogenesis was a primary malformation of body wall closure, with abnormal fusion of the amnion, which had occurred in the first month of development.[6]

Omphalocele and gastroschisis are the 2 most common types of abdominal wall defects. Differentiation between these 2 conditions are shown in table 1.

Table 1. Essential features of omphalocele and gastroschisis

clinical features	Omphalocele	Gastroschisis
Definitions	a congenital defect of the anterior abdomial wall characterized by the protrusion of the intestine covered by a vascular sac composed of peritoneum and amniotic membranes.	a congenital defect of the anterior abdominal wall characterized by the protrusion of the intestine uncovered by the sac.
Incidence	1:4000-1:6000	1:10,000-1:12,000
pathogenesis	an abnormal development of the gut prior to third week of embryonic life which prevents the later return of the midgut into the abdominal cavity.	antenatal or perinatal herniation at the base of the umbilical cord allowing variable amount of intestine to herniate out into the amniotic fluid.
clinical features -umbilical cord	-inserted into the apex or base of the sac.	-the herniated mass is usually to the right of umbilical cord separated by a band of skin.
-herniated content	-small intestines usually plus liver and spleen. (when the liver is involved an omphalocele should be suspected).	-small bowel with not often a portion of the liver. (When bowel alone is seen herniated outside the abdomen gastroschisis should be first choice)
-bowel loop	-usually normal.	-thickening, fibrosis, shortened, frequent by infarcted and atretic.
-abdominal cavity	-less developed than in gastroschisis.	-more developed than omphalocele
-amniotic fluid	-Polyhydraminos usually present. But not always severe.	-may associated with polyhydramnios later.
-associated anomalies (30-70%)	1. Congenital heart defect ■ 33% TOF ■ 19% ASD (secundum) ■ rare VSD, PS, Coarctation, PDA, A-V canal 2. genetic syndrome ■ trisomy D, E, 21 ■ Beckwith-Wiedemann syndrome	-infrequent (14%) jejunoileal malformation.

Though the 2 entities have embryologically difference, differentiation between ruptured omphalocele and gastroschisis is not always possible.[1-2] Detection of membrane may be difficult when not outlined by ascites, however the mass appears contained rather than floating freely in the amniotic fluid, which is suggestive of omphalocele. Body defects are a constant feature of the LBWC (92-96%),[3,4,7] and generally large involvement both the thorax and abdomen. Left sided body wall defect are three times more common than right sided defects. Typically, the herniated organ forms a complex bizarre -appearing mass covered with membranes.

Sonography has an important role in the detection, evaluation and follow up of fetus identified with an anterior abdominal wall defect. In addition to gastroschisis and omphalocele other malformation that should be considered including the LBWC, amniotic band syndrome and cloacal exostrophy. It is important to establish correct diagnosis prenatally and to identify major complex anomalies. In spite of excellent results can be expected following surgical correction of an isolated abdominal wall detect, any how pregnancy termination may be elected when a complex malformations are present.

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