Thoracoschisis associated with diaphragmatic hernia in a 31-week-old stillbirth

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Thoracoschisis is a very rare congenital anomaly and is frequently found with other congenital defects the of limbs and the abdominal wall as a part of limb-body wall complex (LBWC). Early vascular disruption, amnion rupture and intrinsic embryonic maldevelopment are related to the pathogenesis of this complex.

We present a case of thoracoschisis with ipsilateral diaphragmatic hernia. This case had none of the associated anomalies which are seen in LBWC. By ultrasonography performed at 31 weeks' gestation the fetus was misdiagnosed as gastroschisis. In this report we discuss the pathogenetic mechanism of LBWC and our case.

Key words: thoracoschisis, gastroschisis, prenatal ultrasonographic diagnosis, limb-body wall complex.

Thoracoschisis is a very rare congenital anomaly characterized by the herniation of the lung through a defect of the thoracic wall. Most previously reported cases of thoracoschisis and thoraco-abdominoschisis are found in association with other anomalies, most frequently exencephaly/encephalocele with or without cleft lip and/or limb defects, although other defects can also be present[1,2]. Three pathogenic mechanisms have been proposed for this disorder: amnion rupture, vascular disruption and embryonic malformation[3]. Recently it has been hypothesised that limb-body wall complex (LBWC) is a very heterogeneous group of disorders. Some cases show amniotic bands while others demonstrate evidence of vascular disruption or early embryonic maldevelopment[4].

Here we present the clinical, radiological and postmortem findings of a case of thoracoschisis without any other components of the LBWC, and we emphasize the importance of the differential diagnosis between gastroschisis and thoracoschisis by prenatal ultrasonography.

Case Report

A 25-year-old healthy primigravida, a medical doctor working as a general practitioner, was first examined in the 17th week of gestation. Her history revealed a normal pregnancy to that stage. The woman was married to a 28-year-old man, and the couple was nonconsanguineous. Routine obstetric ultrasonography at week 17 was normal. However, ultrasonography at week 31 showed that part of the fetal intestine and almost all the liver were herniated through an abdominal wall defect (Fig. 1). The diagnosis of gastroschisis was made at another hospital. The woman was then referred to our center. The high level of alpha-fetoprotein (410 ng/ml) in the maternal serum at the same week of the diagnostic ultrasound exam was consistent with the fetal morphological findings. There was no history of drug ingestion during pregnancy.

A few days later, the fetus died in utero and labor was induced with an oxytocin infusion. Postmortem examination revealed a female fetus whose weight and length were 1,850 g and 45 cm, respectively. The liver and parts of the intestine and colon were herniated through a right thoracic defect of 4 x 3 cm. The margins of the defect were smooth. Otherwise, the general physical examination was normal (Fig. 2). A total body X-ray showed incomplete formation of the right ribs toward the ventral surface of the chest (Fig. 3). The second and fourth ribs, which exhibited the defect, were separated

cranially and caudally, respectively, in a configuration that allowed the viscera to prolapse. All the other parts of the skeleton and a cranial computed tomography scan were normal. There was also a right-anterolateral diaphragmatic defect of 2 x 3 cm. Unfortunately, no information was available regarding the fetal membranes. The fetus had no adherent bands or membranes. All other pathological exam findings were normal. Microscopic examination of the herniated liver and intestines revealed coagulation necrosis. The placenta was normal with a paracentrally located cord. Efforts to obtain a peripheral blood culture for chromosomal analysis were unsuccessful, but both parents' karyotypes were normal.

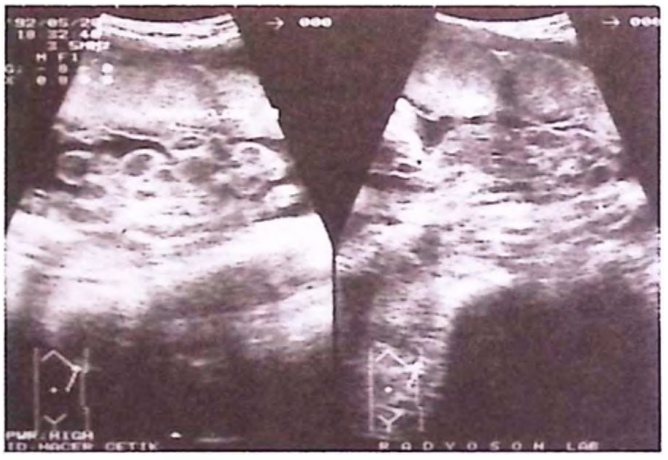

Fig. 1. The ultrasonographic appearance of the fetus demonstrated herniated fetal intestinal loops and the liver.

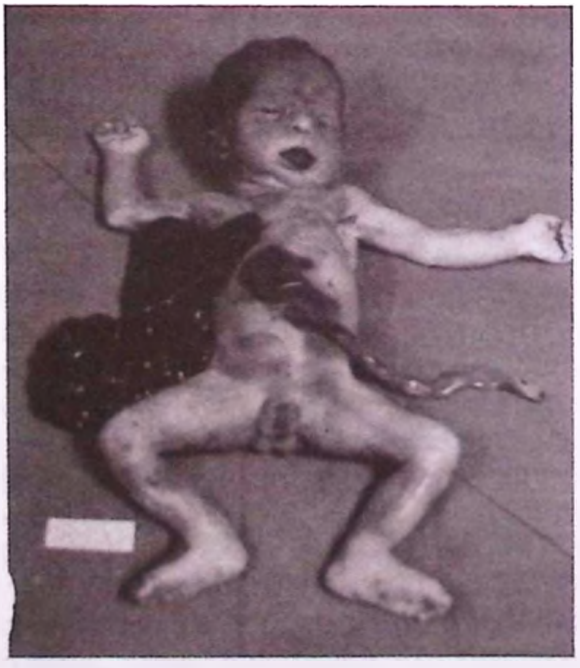

Fig. 2. Macroscopic appearance of the fetus showed no external malformation except right thoracic defect.

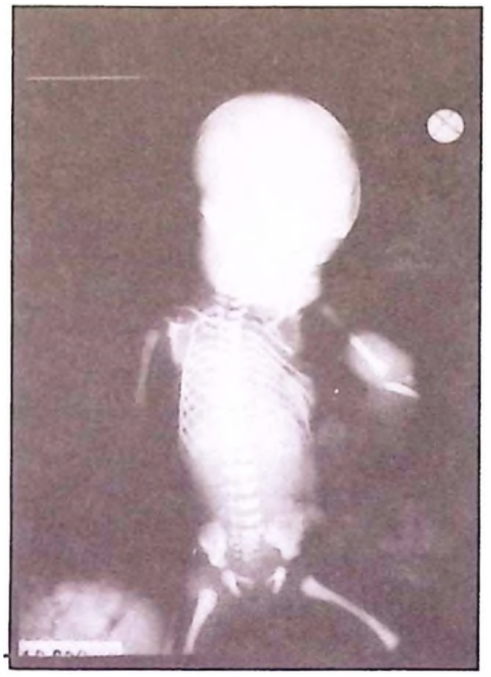

Fig. 3. Postmortem X-ray of the case showed incomplete formation of the right ribs.

Discussion

Thoracoschisis is a very rare congenital anomaly that involves herniation of the lung through an opening in the thoracic wall[1]. Most cases of thoracoschisis reported in the literature are associated with other anomalies such as exencephaly/encephalocele with or without cleft lip and palate, and congenital amputation of the extremities or fingers, which are components of the limb-body wall complex[1-3]. LBWC refers to a variable spectrum of congenital defects that includes abdominal and/or thoracal wall defect with limb and visceral anomalies. There are three main pathogenetic mechanisms related to this complex: amnion rupture, vascular disruption and embryonic maldevelopment[4]. Van Allen et al.[2] proposed LBWC resulted from vascular disruption. Vascular disruption refers to structural anomalies resulting from damage to or interruption of normal embryonic or fetal development of the arteries and veins. The types of structural anomalies that result from vascular disruption are determined by the timing in gestation, and the severity and location of tissue damage[3]. Higginbottom et al.[5] hypothesized that these cases were due to early amniotic rupture and they named the defect Amniotic Band Disruption Sequence. Amnion Rupture Sequence is a disruption complex characterized by rupture of the amnion with secondary effects on the fetus

producing malformations due to interruption of normal morphogenesis, deformations due to distortion of established structures, or mutilations of structures already formed[5,6]. But these theories cannot explain the severe visceral and placental anomalies seen in LBWC. Alternatively, Russo and others[7] have presented evidence that some cases of LBWC result from primary embryonic maldevelopment. Recently, Craven et al.[4] have hypothesized that there are subsets of LBWC that have similar structural abnormalities and a common pathogenesis. They reported five cases of LBWC. Perinatal autopsies of these cases suggested that a primary malformation had occurred. They proposed that these cases represented a pathogenetic subgroup of LBWC.

It has been suggested that vascular disruption is a possible cause in the pathogenesis of amnion disruption sequence, LBWC, gastroschisis, structural anomalies of the limbs, and the Poland, Moebius, and Klippel-Feil sequences[3]. The type of the anomaly is thought to depend on the severity or location of the vascular disruption and on the stage of pregnancy. For instance, while disruption of the embryonic capillary plexus might cause early amnion disruption sequence and limb reduction anomalies, structural anomalies such as Poland, Moebius, and Klippel-Feil sequences are thought to be due to premature ablation of embryonic vessels[3]. The Poland anomaly is a congenital chest wall deformity that involves unilateral aplasia or hypoplasia of the pectoralis major and/or minor muscles, absence or hypoplasia of the nipple and breast, and deformity or absence of ribs and/or hands. The subclavian artery supply disruption sequence has been suggested as a factor in the pathogenesis of the Poland anomaly[8]. Gastroschisis, which generally appears as an isolated anomaly, or infrequently together with certain anomalies related to the intestines, has been attributed to the ablation or occlusion of the omphalomesenteric artery[3,9].

The relatively localized anomaly in our fetal patient may also have been caused by an embryonic or fetal vascular disruption. Based on the laterality of the defect and the lack of any associated anomaly other than an ipsilateral congenital diaphragmatic hernia, we believe that the pathogenetic mechanism behind the problem in this fetus was similar to that behind gastroschisis. Specifically, we conclude that this case of thoracoschisis with congenital diaphragmatic hernia was caused by premature ablation or occlusion of a specific artery.

This case also illustrates the importance of differentiating thoracoschisis from gastroschisis on ultrasonography. When the intestinal loops appear as cystic structures in the amniotic fluid, the diagnosis of gastroschisis is made easily by prenatal ultrasonography[10]. However, our case involved thoracoschisis with congenital diaphragmatic hernia, which can produce images similar to those of gastroschisis. Also, most reported cases of thoracoschisis have been associated with other anomalies. This differential diagnosis is important with regard to genetic counseling and appropriate postnatal management of the fetus.

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