CASE REPORT

Eventration of the Diaphragm in Trisomy 18: Providing Information and Helping Choose a Treatment and Management Plan with an Exact Diagnosis

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Abstract

Trisomy 18 is one of the most common aneuploidies, and its prognosis is controversial due to the low survival rate associated with it. Thus, intensive management and treatment strategies for this genetic trisomy syndrome are essential. Because patients with trisomy 18 exhibit severe neurodevelopmental delay in long-term survival, treatment strategies can be complex and difficult. This study describes a complex case of trisomy 18 with rapid changes and complications in the third trimester. These complications made differentiation between diaphragmatic hernia and diaphragmatic eventration on ultrasound difficult. Since this case was associated with rapid changes and required rapid delivery, close examination was not possible. Further, it is difficult to provide information and help decide whether to intervene without an exact diagnosis. In conclusion, an interdisciplinary team approach, including parental involvement, is vital for helping families to make appropriate decisions. For this reason, an accurate ultrasound diagnosis is necessary.

Keywords: Eventration of the diaphragm, Perinatal care, Trisomy 18.

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Introduction

As an aneuploidy that is both common and severe, trisomy 18 has a controversial prognosis. Infant mortality is high in this condition, with approximately 50% of babies with trisomy 18 living longer than 1 week and 5–10% of children living beyond the first year.[1] Management with treatment strategies that encompass neonatal care, genetic counseling, and medical ethics is clinically essential for this genetic trisomy syndrome.

Patients with trisomy 18 who survive in the long term exhibit severe neurodevelopmental delay,[2] making treatment difficult and complex. Therefore, an interdisciplinary team approach that includes parental involvement is vital for helping families of patients with trisomy 18 make appropriate decisions.

Here, we report a case of unusual fetal presentation of eventration of the diaphragm caused by trisomy 18.

Case Description

A 39-year-old pregnant woman, gravida three para two, presented to our perinatal center at 27 weeks of gestation, having been referred from a private clinic due to a diagnosis of fetal growth restriction during a routine pregnancy checkup. Ultrasound showed normal amniotic fluid volume, intrauterine growth restriction with an estimated fetal weight of 817 g (–1.6 standard deviation), mega cisterna magna, cerebellar hypoplasia, ventricular septal defect (VSD), and an abnormal location of the ductus *venosus* (Figs 1A to C).

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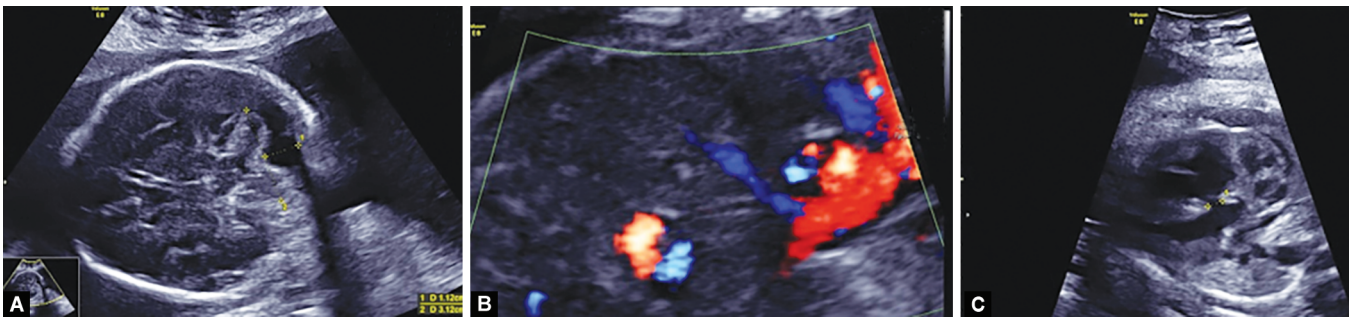
In general, private clinics in Japan, the second trimester fetal morphology assessment is usually attempted by general OB/GYN doctors, who are not specialized in ultrasonography. Conversely, the first trimester fetal screening is provided in perinatal centers, general hospitals, or some private clinics where scanning is performed by perinatal ultrasound specialists. In this case, the second trimester morphology assessment was performed, but no anomalies were detected.

Since the fetal growth restriction was associated with congenital anomalies, amniocentesis was conducted. Chromosomal analysis revealed 47, XY, +18. After genetic counseling was provided by us and pediatricians, the mother and her family chose to continue with the necessary medical



Figs 1A to C: Ultrasound images of the fetus at 27 weeks of gestation: (A) Mega cisterna magna and cerebellar hypoplasia; (B) ventricular septal defect; (C) Abnormal course of ductus *Venosus*

care for their child, requesting medical support for the fetus and neonate. The clinical course was uneventful in both the mother and the fetus, except that polyhydramnios occurred after 35 weeks of gestation.

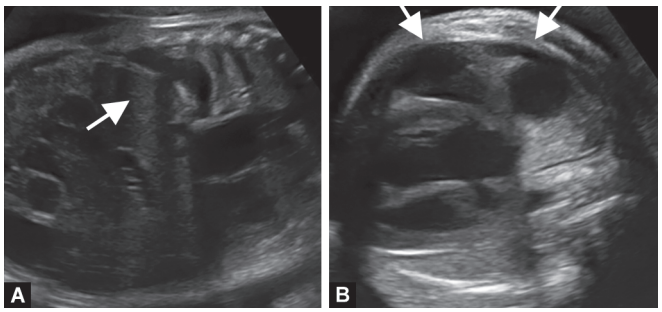
At 39 weeks of gestation, fetal ultrasound showed that the right lung was becoming rapidly decompressed due to entrapment of the right lobe of the liver in the right thoracic cavity (Fig. 2A). An anechoic cyst lesion was also found in the thoracic cavity (Fig. 2B). In addition, polyhydramnios was present, and cardiac contractions were weakened due to increased intrathoracic pressure. A diagnosis of circulatory failure associated with diaphragmatic hernia or diaphragmatic eventration was also made.

The couple and their family confirmed their requests for continued medical treatment for the fetus after learning of the ultrasound results. Therefore, induced delivery was planned. The mother delivered vaginally, with the infant weighing 2200 gm and having an Apgar score of 4/6 (1/5 min). Intubation and resuscitation were performed in the delivery room before admittance to the neonatal intensive care unit. Neonatal examination revealed a VSD, cerebellar hypoplasia, rocker-bottom feet, and right hydronephrosis. Chest radiography showed elevation of the diaphragm without air findings in the thorax; therefore, a diagnosis of congenital diaphragmatic eventration was considered. Ultrasound showed that the congenital diaphragmatic eventration improved with conservative treatment at 11 days of age, and the artificial respirator was withdrawn. The patient underwent pulmonary artery banding for the VSD at 19 days and was discharged at 4 months of age; thereafter, the patient received home oxygen therapy.

Discussion

In the present case, we struggled with differentiation between diagnoses of diaphragmatic eventration and congenital diaphragmatic hernia (CDH) because both anomalies can be associated with ultrasonographic findings in the thorax. However, the prognoses of these two anomalies are extremely different.

Congenital diaphragmatic hernia is a congenital anomaly of the diaphragm, with pulmonary hypoplasia and persistent pulmonary hypertension as consequences. Surgical repair is necessary after stabilization of cardiorespiratory function.



Figs 2A and B: Ultrasound image of cystic lesion in the thorax before delivery: (A) Coronal view showing the right lobe of the liver was entrapped in the right thoracic cavity; (B) Horizontal view showing cyst-like image in the thoracic cavity

Of infants diagnosed with CDH that underwent surgery, 92% survived.[3] The mortality rate for infants with CDH is 37%.[4,5]

By contrast, diaphragmatic eventration does not always require surgery. Only 23% of patients with congenital diaphragmatic eventration are symptomatic and require surgical repair.[6] The mortality rate in cases of diaphragmatic eventration is only 1–8%.[7]

Because the prognoses of CDH and diaphragmatic eventration are quite different, accurate diagnosis by ultrasound is required. This can help to determine the treatment plan for the patient. However, in this case, fetal ultrasound showed liver entrapment in the right thoracic cavity with one decompressed lung and an anechoic cystic lesion in the thoracic cavity with polyhydramnios, making it difficult to distinguish between CDH and diaphragmatic eventration. Without an accurate diagnosis, perinatal counseling, in this case, became more complex.

Among children with trisomy 13, 5-year survival was 9.7%; among children with trisomy 18, it was 12.3%. Therefore, careful provision of information and in-depth discussion is needed during genetic counseling between various specialties of medical staff and the pregnant women and her family. We believe that aggressive medical intervention for these children has been recommended in recent years. However, the natural duration of life for patients with trisomy 18 is dependent upon complications associated with chromosomal abnormalities. For example, infants with trisomy 18 and omphalocele have been shown to have a 50%

increased risk of mortality at 1 and 5 years compared to those without an omphalocele.[8]

Leonardo et al. have also reported the case of a 12-year-old girl diagnosed with full trisomy 18 and multiple malformations, including Dandy-Walker syndrome and congenital heart defects, as an example of long-term survival.[9]

The disciplines of pediatric genetics, cardiology, and cardiac surgery have changed and grown immensely in the last 50 years and continue to do so.[10] With advancing medical treatment, survival rates of children with trisomy 13 and 18 have increased and life spans have become longer. Thus, among infants born with trisomy 13 or 18, we should assume that more and more parents may decide that they want life-sustaining treatment. Furthermore, parents will likely have accessed digital media resources and be aware of the possibility that their infant can survive for years and be accepted as a loved and loving family member. Given these realities, it seems difficult to ethically justify a physician's refusal to provide life-sustaining treatment to such infants.[11] As more syndromes, diseases, interventions, and procedures are discovered, there are both ongoing and new ethical considerations to be debated and discussed. As illustrated by this case, determining the benefits that a complex surgery would provide an infant with trisomy 18 is difficult because the range of possible comorbidities is so wide that there is uncertainty over the child's life expectancy. The amount of suffering a parent is willing to put their child through for both quality and quantity of life also varies among families. Therefore, there are significant parental decisions to be made following prenatal diagnosis of trisomy 18.

Some parents may occasionally choose artificial abortion upon learning that their fetus is affected by trisomy 18. Alternatively, parents may also choose palliative care, expectant management, or full intervention. Because information based on an inaccurate ultrasound diagnosis can result in loss of life, the possibility of sudden changes must always be considered. Since this case was associated with rapid changes and required rapid delivery, close examination was not possible; therefore, ultrasound diagnosis and provision of information were essential.

In conclusion, during genetic counseling for parents of fetuses diagnosed with trisomy 18, we must provide realistic outcome data to assist families with decision-making. Therefore, it is necessary to diagnose and estimate prognosis as accurately as possible. For these reasons, an accurate ultrasound diagnosis is necessary; it is difficult to provide information and help choose a treatment and management plan without an exact diagnosis.

Acknowledgments

Ethical approval and informed consent: Informed consent was obtained from the patient. Although the Institutional Review Board (IRB) of St Marianna University Hospital does not require IRB approval in the present case, the confidentiality of the patient involved was protected. All patient records and information were anonymized and de-identified prior to analysis.

Data availability: The data related to this study were collected from medical records, paying attention to confidentiality.

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