Clinical Case Report

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Congenital diaphragmatic hernia in association with congenital short esophagus

A case report

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Abstract

Rationale: Congenital diaphragmatic hernia (CDH) associated congenital anomalies are present in about 25%. Congenital short esophagus (CSE) is a relatively rare condition. Both CDH and congenital intrathoracic stomach caused by CSE can be diagnosed in utero. However, CSE can be easily misdiagnosed in utero.

Patient concerns: We present a case of left CDH with CSE in a female neonate who was diagnosed at 24 weeks gestational age by antenatal ultrasound.

Diagnoses: The neonate with CDH and congenital intrathoracic stomach due to CSE was confirmed by operation. Gastroesophageal reflux disease (GRED) occurred after operation.

Interventions: The left diaphragm was repaired, and gastric fixation by gastropexy, gastric folding anti-reflux procedure and operation of longitudinal incision and transverse suture for pyloroplasty procedure was underwent. Gastroesophageal reflux disease (GRED) occurred after operation and jejunal tube feeding was placed.

Outcomes: This patient is currently alive 12 months post-operation with GRED.

Lessons: To our knowledge, this is the first documented case of this rare type of CDH combined with congenital intrathoracic stomach caused by CSE. This condition could not be surgically repaired due to the extremely short esophagus. Early recognition of intrathoracic stomach associated with CSE is important as it is associated with difficult management and significant postnatal complications. The prognosis is cautiously guarded, and the parents should be appropriately counseled.

Abbreviations: CDH = congenital diaphragmatic hernia, CSE = congenital short esophagus, CT = computed tomography, LHR = lung-to head ratio, MRI = magnetic resonance imaging, US = ultrasonography.

Keywords: congenital diaphragmatic hernia, congenital short esophagus, diagnosis, intrathoracic stomach

1. Introduction

Congenital diaphragmatic hernia (CDH) is one of the more common congenital anomalies with its incidence as one in 2200 live births. The defect in the diaphragm allows abdominal organs to protrude into the chest creating a mass effect that impedes lung development during a critical stage of normal lung development. CDH is usually diagnosed in utero by means of routine ultrasound analysis, and the most utilized prenatal predictors are evaluated by measuring the lung volume, including the lung–head ratio (LHR), and observed to expected LHR (O/E LHR).[1] Associated congenital anomalies are present in about 25%. Early recognition of lethal anomalies are essential to the prenatal counseling process and postnatal management planning.

Congenital short esophagus (CSE) is a relatively rare condition that is associated with secondary intrathoracic stomach. The clinical misdiagnosis rate was high. The suggestive features include congenital short esophagus, intrathoracic stomach, and segmental arterial supply to the stomach from the thoracic aorta, absence of left gastric artery extension across the diaphragm into the thorax and absence of a hernia sac.[2]

Cases of CDH combined with congenital intrathoracic stomach caused by CSE are less seen. To our knowledge, this is the first case documented case of this rare type of CDH combined with congenital intrathoracic stomach caused by CSE in English literature. Knowledge concerning the diagnosis and management of CCAM with CSE is reviewed.

2. Case report

A female infant, with a prenatal diagnosis of CDH, was born at 34 week gestation via cesarean section (birth weight, 1760 g). She was initially diagnosed with a mild left CDH at 24 weeks

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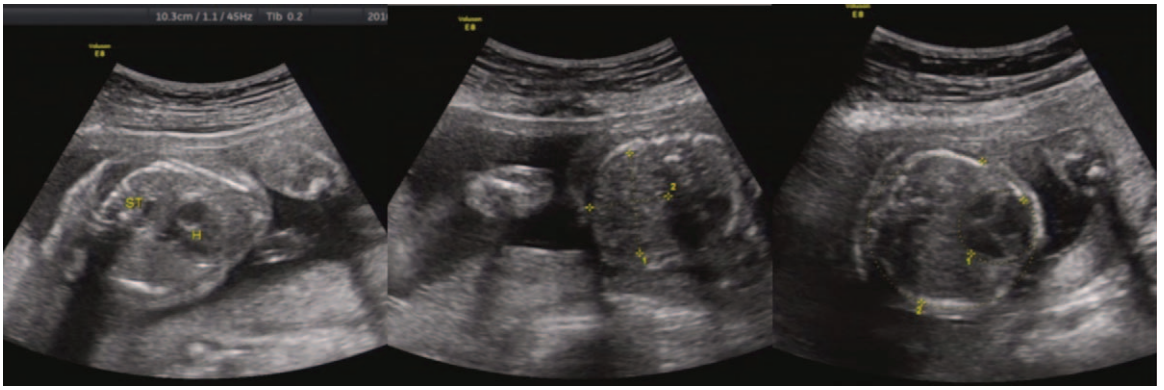

Figure 1. Antenatal ultrasound revealed a mild left CDH with bowel and stomach herniation, a LHR of 1.69, an O/E LHR of 60%. CDH = congenital diaphragmatic hernia, LHR = lung-to head ratio.

gestational age. Antenatal ultrasound revealed a CDH with bowel, stomach and liver herniation, a lung-to head ratio (LHR) of 1.69, an O/E LHR of 60% (Fig. 1). There was no prenatal intervention. Amniocentesis was performed, but karyotyping showed no distinct abnormality. Maternal blood test results were all negative for active infection by adenovirus, cytomegalovirus, or toxoplasma. Apgar scores were 7, 8, and 10 at 1, 5, and 10 minutes, respectively. She was supported with mechanical ventilation. She did not require extracorporeal membrane oxygenation and inhaled nitric oxide.

On admission, it is difficult to insert a gastric tuber. Upper gastrointestinal radiography showed abdominal organs protruding into the thoracic cavity, heart displacement and stomach was in the right thoracic (Fig. 2). On the third day of life, she had remained relatively stable and underwent operation. During surgery, a large diaphragmatic defect was identified, the size of defect is about 5 cm × 4 cm, which allowed all the small intestine, most of the colon and spleen into the thoracic cavity. After content of will hernia content pushes an abdominal cavity and repaired the left diaphragm, the major intraoperative finding was herniation of the entire stomach into the posterior mediastinum. The gastric blood supply was found to arise from anomalous blood vessels originating from the thoracic rather than abdominal aorta. No hiatal hernia sac could be identified. CDH and CSE with intrathoracic stomach were diagnosed, gastric fixation by gastropexy, gastric folding antireflux procedure and operation of longitudinal incision and transverse suture for pyloroplasty procedure was underwent. Gastroesophageal reflux disease (GRED) occurred after operation and jejunal tube feeding was placed. The patient is currently alive 12 months postoperation with GERD (Fig. 3).

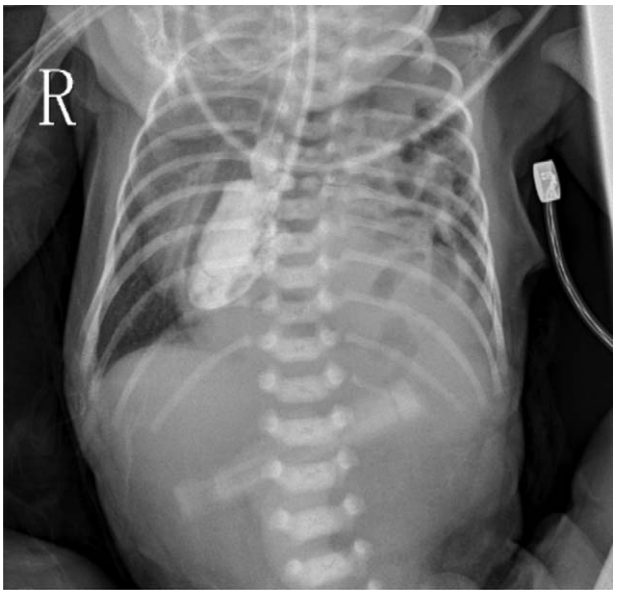

Figure 2. Upper gastrointestinal radiography showed abdominal organs protruding into the thoracic cavity, heart displacement, and stomach was in the right thoracic.

3. Discussion

CDH combined with CSE is an extremely rare condition. To our knowledge, there is the first report of CDH combined with intrathoracic stomach due to short esophagus in a preterm infant in English literature. In the present case, CSE was misdiagnosed as CDH causing intrathoracic stomach.

Congenital intrathoracic stomach is defined as a condition in which at least two-third of the stomach is herniated into the posterior mediastinum, and can be the result of developmental abnormalities, including CDH, congenital hiatal hernia, CSE and heterotopic mucosa of the lower portion of the esophagus.[2] The commonest cause of a neonatal intrathoracic stomach is CDH, which is one of the more common congenital anomalies with a frequency of 1/2200 live births.[3,4] Associated congenital anomalies are present in about 25%. However, the combination described above of congenital short esophagus with intrathoracic stomach is extremely rare. To our knowledge, there was no report in the literature of CDH combined with intrathoracic stomach due to short esophagus in a preterm infant.

CDH is a congenital defect in the diaphragm that allows herniation of abdominal contents into the fetal chest, and also can be suggested antenatal by the absence of a stomach bubble on ultrasound.[4,5] Hence, although prenatal ultrasound investigation revealed that stomach bubble was shown in the chest, CDH was diagnosed. CSE is a rare and serious congenital development malformation that is associated with secondary intrathoracic stomach. The clinical misdiagnosis rate was high. The suggestive features include congenital short esophagus, intrathoracic stomach, segmental arterial supply to the stomach from the thoracic aorta, absence of left gastric artery extension across the diaphragm into the thorax and absence of a hernia sac.[2,6]

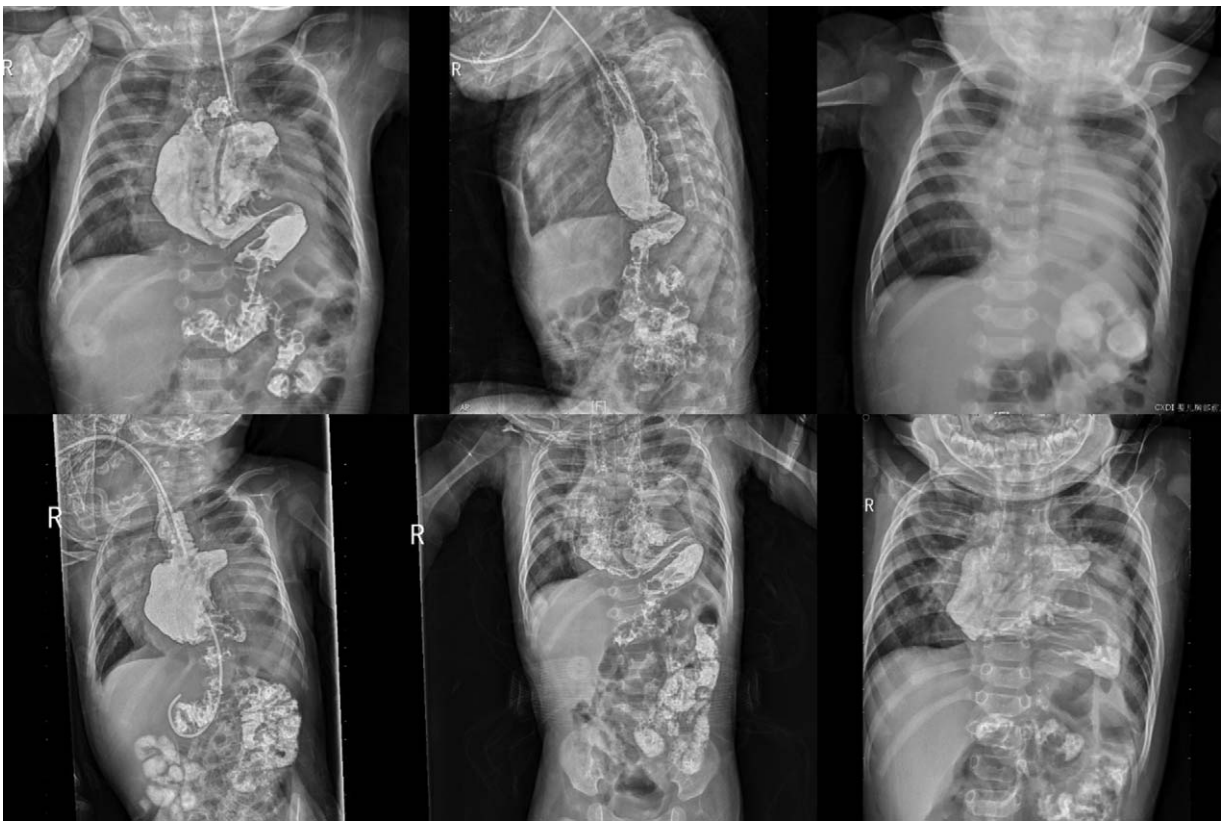

Figure 3. A1, A2, B, C, D, E: Upper gastrointestinal radiography were performed at postoperative 2, 4, 5, 6, and 12 months showing intrathoracic stomach.

For CDH, an oro- or nasogastric tube was routinely placed in order to prevent bowel distension and any additional ipsilateral lung compression. In this case, we found it is difficult to insert stomach tube. Congenital esophageal atresia was first considered, Upper gastrointestinal radiography showed stomach was in the right thoracic. Based on the principle that all conditions were explained by 1 disease, CDH was the first factor to consider. However, during surgery, CDH combined with congenital intrathoracic stomach due to short esophagus was supplement diagnosed. As the imagological examination of CDH and CSE are stomach in the chest, it is easy misdiagnosis, need to be confirmed by surgery. In this case, although the patient is currently alive 12 months postoperation with GERD, her condition could not be surgically repaired due to the extremely short esophagus, which is about 5 cm long. Herein, early recognition of intrathoracic stomach associated with CSE is important as it is associated with difficult management and significant postnatal complications. The prognosis is cautiously guarded, and the parents should be appropriately counseled.

Thus, detailed antenatal assessment to differentiate between CDH and CSE is crucial. Characteristics of an intrathoracic stomach on ultrasound in utero include a hypoechogenic lesion in the posterior mediastinum with no evidence of mediastinal shift, and absence of the stomach bubble.[2] Also in CDH, an intraabdominal stomach bubble may be intermittently present as the sliding movement of the stomach body through the hiatal orifice, however in CSE, such movement is restricted by the short length of the esophagus and the stomach bubble would consistently be absent in repeated antenatal imaging.[2] Then, magnetic resonance imaging (MRI) may be a useful tool to delineate the exact location of the esophagogastric junction.[2]

4. Conclusion

In summary, we report this rare case of CDH combined with congenital intrathoracic stomach caused by CSE. This report describes a truly rare case, and her condition could not be surgically repaired due to the extremely short esophagus. Early recognition of intrathoracic stomach associated with CSE is important as it is associated with difficult management and significant postnatal complications. Fetal series ultrasound and MRI may be useful in this situation. The prognosis is cautiously guarded, and the parents should be appropriately counseled.

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