



Prenatal findings of serpentine-like syndrome with congenital intrathoracic stomach: differential diagnosis from congenital diaphragmatic hernia

Kazuya Mimura¹ · Masayuki Endo¹ · Keiko Matsuoka² · Takuji Tomimatsu¹ · Yuko Tazuke³ · Hiroomi Okuyama³ · Makoto Takeuchi² · Tadashi Kimura¹

Received: 1 June 2018 / Accepted: 20 July 2018 / Published online: 9 August 2018

© The Japan Society of Ultrasonics in Medicine 2018

Abstract

Congenital rachischisis and brachioesophagus with secondary intrathoracic stomach, which is described as “serpentine-like syndrome”, is a very rare condition. We report the prenatal findings of serpentine-like syndrome in a fetus at 18 weeks of gestation. The stomach was entirely elevated into the thoracic area, but the liver retained its normal position. Our initial diagnosis was isolated left congenital diaphragmatic hernia (CDH). At 20 weeks of gestation, the pregnancy was terminated, and a male newborn weighing 338 g was vaginally delivered. The autopsy findings revealed an extremely short esophagus, and the stomach was located in the posterior mediastinum as a result of congenital esophagus hiatal hernia. A severe case of rachischisis was identified from the cervical to thoracic vertebrae. The prognosis of congenital intrathoracic stomach associated with serpentine-like syndrome is extremely poor compared to the prognosis of isolated CDH. It is difficult to make a definite diagnosis by ultrasound alone; however, atypical findings, such as the presence of the entire intrathoracic stomach despite the normally located liver, may warrant further exploration. Obstetricians and neonatal clinicians presented with similar cases should be alerted to the significance of these prenatal findings and the differences in outcomes to ensure appropriate counseling of parents.

Keywords Congenital hiatal hernia · Congenital diaphragmatic hernia · Intrathoracic stomach · Prenatal diagnosis · Serpentine-like syndrome

Introduction

Congenital rachischisis and brachioesophagus with secondary intrathoracic stomach is a very rare condition, and only a small number of cases have been previously reported [1–7]. This condition has been described as “serpentine-like syndrome” because it creates an anatomy similar to that of a snake [1, 2]. Here, we report the prenatal ultrasonographic

findings of serpentine-like syndrome resembling an isolated left congenital diaphragmatic hernia (CDH) in a fetus. The stomach was entirely elevated into the thoracic area, but the liver retained its normal position. We are not aware of any previously reported case of serpentine-like syndrome with congenital intrathoracic stomach resembling isolated CDH. This condition was difficult to differentiate from isolated CDH based only on prenatal ultrasonographic findings.

✉ Kazuya Mimura
kazuya.med_ob-gyne@hotmail.co.jp

¹ Department of Obstetrics and Gynecology, Osaka University Graduate School of Medicine, 2-2 Yamadaoka, Suita, Osaka 565-0871, Japan

² Department of Pathology, Osaka Women’s and Children’s Hospital, Izumi, Osaka, Japan

³ Pediatric Surgery, Osaka University Graduate School of Medicine, Suita, Osaka, Japan

Case report

A 35-year-old woman (gravida 3, para 0) was referred to our hospital at 18 weeks of gestation for evaluation of an echo-free mass in the fetal thorax. There had been no complications in the pregnancy, and there was nothing out of the ordinary in the patient’s family history. A prenatal ultrasound of the transverse section of the fetal thorax revealed a tubular cystic structure of simple echogenicity,

which suggested the stomach was located behind the heart. Thus, CDH with intrathoracic stomach was suspected (Fig. 1). The heart appeared to be located to the right, and the lungs appeared to be small, with a lung-to-thorax transverse area ratio (L/T ratio) of 0.07. The observed/expected lung area-to-head circumference ratio (O/E LHR) was 31%. In the sagittal section, the liver was normally located below the diaphragm. No other congenital abnormalities were detected. No modification of the cystic structure was observed during the ultrasound. One possible diagnosis was isolated left CDH with protrusion of the stomach into the thorax. The patient and her family received counseling for suspected CDH and the likely postnatal complications, and at 20 weeks of gestation the patient was admitted to terminate the pregnancy. A newborn male weighing 338 g was vaginally delivered using intravaginal gemeprost. The head appeared to be sitting directly on the shoulders, but no other external abnormality was observed (Fig. 2). A chorionic villi culture for postnatal karyotyping revealed a normal karyotype (46, XY). Assessment of the fetal anatomy by autopsy revealed dextrocardia with right mediastinal shift. There was no cardiac abnormality. In addition, both lungs were hypoplastic with a lung-to-body weight ratio of 0.013. The esophagus was extremely short, and the stomach was located in the posterior mediastinum because of a congenital esophagus hiatal hernia (Fig. 3). There was no defect in the diaphragm. The spleen, pancreas, and part of the duodenum were also located in the posterior mediastinum esophageal hiatus. The liver, kidneys, and adrenal glands were in their normal positions. There was no intestinal malrotation. A severe case of rachischisis was observed from the lower cervical to upper thoracic vertebrae (Fig. 4). The final diagnosis was serpentine-like syndrome based on the autopsy findings.



Fig. 1 Transverse view of the prenatal ultrasound shows an intrathoracic stomach and a right-shifted heart



Fig. 2 Postmortem image showing the appearance of the head sitting directly on the shoulders

Discussion

The exact cause of serpentine-like syndrome is unknown, but it is believed to occur in very early stages of gestation. The primary defect of this syndrome is underdevelopment of the esophagus, which causes the stomach to develop high



Fig. 3 Autopsy findings show that the stomach (arrowhead) is localized in the thorax. The arrow highlights the short esophagus



Fig. 4 Anteroposterior X-ray shows rachischisis of the cervical spine

in the thorax. In addition, rachischisis occurs in the cervical vertebrae [3, 8].

Because of the right-shifted heart with intrathoracic stomach and the lack of other anomalies, our initial diagnosis was isolated left CDH. Although isolated CDH is a life-threatening condition due to lung hypoplasia that leads to persistent pulmonary hypertension, the prognosis of CDH with intrathoracic stomach is not always poor. Previous studies have attempted to determine the prenatal prognostic factors of fetal CDH. The best validated tool is the ultrasonographic measurement of lung size and liver position, which can be used to predict lethal pulmonary hypoplasia [9]. In our case, the *L/T* ratio was 0.07, the *O/E LHR* was 31%, and the liver retained its normal position, which is associated with an expected survival rate of 70–80% [10–12]. Conversely, the prognosis of congenital intrathoracic stomach associated with serpentine-like syndrome is extremely poor. Of the eight cases reported in the English literature [1–7], six patients died of respiratory insufficiency soon after birth, and the only two surviving patients were kept alive with long-term feeding via a jejunostomy tube, followed by esophageal elongation. Even when surgical intervention is feasible, the prognosis of serpentine-like syndrome is greatly influenced by the preoperative respiratory stability and nutritional status.

The prenatal ultrasonographic findings of serpentine-like syndrome have not been well described in previous reports. The presence of a cystic mass in the fetal thorax should

alert the sonographer to consider differential diagnoses of congenital chest anomalies such as diaphragmatic hernia or macrocystic adenomatoid malformation of the lung, or rarer conditions, such as esophageal duplication or neurenteric cyst [13]. Congenital hiatal hernia is not commonly considered in the differential prenatal diagnosis of cystic chest anomalies. In our case, the presence of an entire intrathoracic stomach, despite the normally located liver, may be a specific characteristic of serpentine-like syndrome compared to prenatal findings of typical CDH. Because patients with congenital intrathoracic stomach may have a poorer prognosis than those with isolated CDH, it is important to accurately determine the prenatal ultrasonographic findings of CDH and other conditions, such as serpentine-like syndrome. Serpentine-like syndrome is very rare, and it is difficult to make a definite diagnosis by ultrasound alone; however, atypical findings, such as the presence of an entire intrathoracic stomach despite a normally located liver, may warrant further exploration, such as detailed sonographic investigation by experts or fetal MRI. Therefore, obstetricians and neonatal clinicians presented with similar cases should be alerted to the significance of these prenatal findings and the different outcomes to ensure appropriate counseling of parents.

Acknowledgements This study was supported by Grants-in-Aid for Scientific Research (17K11233, 15K10669, 26670724) from the Ministry of Education, Culture, Sports, Science and Technology of Japan, and from the Japan Society for the Promotion of Science. The authors would like to thank Enago (<http://www.enago.jp>) for the English language review.

Compliance with ethical standards

Ethical statements This article does not contain any studies with human or animal subjects performed by any of the authors. Written informed consent was obtained from the patient.

Conflict of interest The authors declare that there are no conflicts of interest regarding the publication of this paper.

References

1. Katz MS, Hess DJ, Caty MG, et al. Of snakes and babies: intrathoracic stomach and vertebral rachischisis. A serpentine-like syndrome? *J Pediatr Surg*. 2008;43:1385–9.
2. Deprez FC, Debauche C, Clapuyt P, et al. Multiorgan developmental anomalies presenting as a variation of the serpentine-like syndrome: cervical fusion and brachioesophagus with intrathoracic stomach and malposition of duodenopancreas and spleen. *J Pediatr Surg*. 2009;44:E25–8.
3. Nakamura H, Okazaki T, Koga H, et al. Congenital brachioesophagus with secondary intrathoracic stomach associated with rachischisis described as “serpentine-like syndrome”: a case report and literature review. *Pediatr Surg Int*. 2012;28:63–6.

4. Lall A, Morabito A, Bianchi A. “Total Gastric Dissociation (TGD)” in difficult clinical situations. *Eur J Pediatr Surg*. 2006;16:396–8.
5. Leung AW, Lam HS, Chu WC, et al. Congenital intrathoracic stomach: short esophagus or hiatal hernia? *Neonatology*. 2008;93:178–81.
6. Dorum BA, Korkmaz S, Özkan H, et al. Serpentine-like syndrome associated with encephalocele. *Clin Dysmorphol*. 2016;25:110–2.
7. Belez-Meireles A, Steenhaut P, Hocq C, et al. “Serpentine-like syndrome”—a very rare multiple malformation syndrome characterised by brachioesophagus and vertebral anomalies. *Eur J Med Genet*. 2017;60:100–4.
8. Pierre R, Eduardo DR, David AP. Pathology of pediatric gastrointestinal and liver disease. New York: Springer; 2004. p. 3–18.
9. Kilian AK, Büsing KA, Schuetz EM, et al. Fetal MR lung volumetry in congenital diaphragmatic hernia (CDH): prediction of clinical outcome and the need for extracorporeal membrane oxygenation (ECMO). *Klin Padiatr*. 2009;221:295–301.
10. Deprest JA, Flemmer AW, Gratacos E, et al. Antenatal prediction of lung volume and in utero treatment by fetal endoscopic tracheal occlusion in severe isolated congenital diaphragmatic hernia. *Semin Fetal Neonatal Med*. 2009;14:8–13.
11. Usui N, Okuyama H, Kanamori Y, et al. The lung to thorax transverse area ratio has a linear correlation with the observed to expected lung area to head circumference ratio in fetuses with congenital diaphragmatic hernias. *J Pediatr Surg*. 2014;49:1191–6.
12. Kitano Y, Okuyama H, Saito M, et al. Re-evaluation of stomach position as a simple prognostic factor in fetal left congenital diaphragmatic hernia: a multicenter survey in Japan. *Ultrasound Obstet Gynecol*. 2011;37:277–82.
13. Di Francesco S, Lanna MM, Napolitano M, et al. A case of ultrasound diagnosis of fetal hiatal hernia in late third trimester of pregnancy. *Case Rep Obstet Gynecol*. 2015;2015:194090.