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Case Report

Congenital paraesophageal hernia with gastric outlet obstruction in a neonate with Cornelia de Lange Syndrome ☆,☆☆

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ABSTRACT

We describe a case of a newborn being treated for encephalopathy and seizures, whose radiographs since the first day of life demonstrate a persistent ovoid lucency over the central lower chest. A CT performed confirmed a type IV hiatal hernia, which is defined as a paraesophageal type hernia containing a portion of the abdominal viscera. This infant's hernia included the distal stomach, pylorus, and proximal duodenum. There was no volvulus or ischemic change at surgery. The patient underwent successful reduction, fundoplication, and gastrostomy placement with hospital discharge after further stabilization of additional medical problems. Genetic testing later confirmed Cornelia de Lange Syndrome Type V, which has been associated with gastrointestinal manifestations and congenital diaphragmatic hernias.

Introduction

This report discusses an uncommon case of neonatal congenital paraesophageal hernia containing a portion of the gastric antrum, pylorus, and proximal duodenum in a patient with Cornelia de Lange Syndrome. Further discussion of the imaging findings and treatment are given with reflections on the complexity of the case.

Case report

A 2215 gm female born at 38w1d via normal spontaneous vaginal delivery was transferred to our institution from an outside hospital for respiratory distress and abnormal posturing with concerns for hypoxic ischemic encephalopathy and seizures. Amniotic fluid was meconium stained, and the neonate was small for gestational age. Physical exam was also notable for dysmorphic facial features with micrognathia and left talipes equinovarus. She initially required positive pressure ventila-

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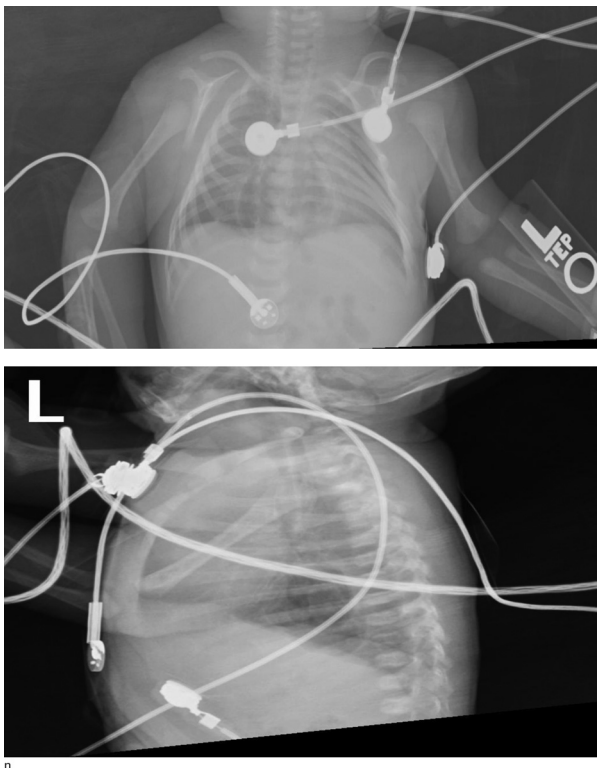

Fig. 1 – Day of life 0. Frontal and lateral radiographs demonstrate lucency over the lower chest suggestive of a hernia.

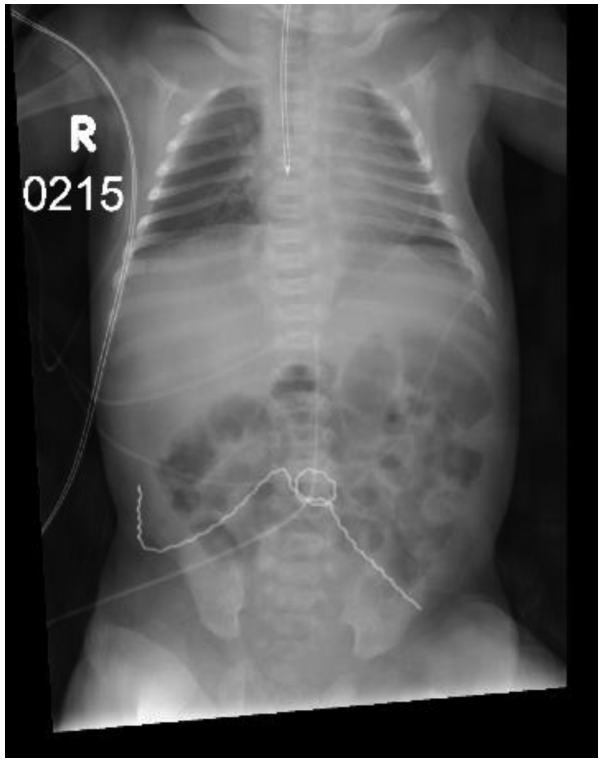

Fig. 2 – Day of life 1. Persistent abnormal mediastinal contour. Esophageal probe and umbilical catheter in place.

tion but was weaned to room air. Initial radiographs on days 0 and 1 of life demonstrated curvilinear lucency over the lower mediastinum suggesting a hiatal hernia (Figs. 1 and 2).

She met clinical and laboratory criteria for the hypothermia protocol. Continuous EEG demonstrated seizures during her hospitalization in the neonatal intensive care unit (NICU), which were treated with antiepileptic drugs. No evidence of hypoxic ischemic injury was demonstrated on MRI. The patient was nil per os (NPO) and was maintained on total parenteral nutrition during the cooling and rewarming protocol with eventual introduction of enteric feeding as well as continuation of parenteral nutrition.

She had new abdominal distension and non-bilious emesis which began a few days after initiating low volume trophic feedings. A radiograph at 7 days of life (Fig. 3) demonstrated increasing gastric distension despite normal positioning of the feeding tube, with a spherical contour of the stomach bubble and a paucity of distal bowel gas. The enteric tube projected over the stomach. A lateral decubitus film was added to exclude free intra-peritoneal gas. A CT of the chest, abdomen, and pelvis was then performed to further evaluate for abdominal pathology and to identify the cause of the abnormality on the radiographs.

CT demonstrated herniation of the gastric antrum, pylorus, and proximal duodenum into the chest, with a dilated gastric lumen (Fig. 4). The position of the gastroesophageal junction was normal. The bowel downstream was decompressed and enhanced normally. Although the findings were likely present since birth, the development of gastric distention was suspicious for developing obstruction.

Physical exam demonstrated palpable fullness and distension in the left upper abdomen, but the abdomen was soft otherwise. The infant had normal laboratory values and vital signs. The patient was taken to the operating room the next morning since her clinical exam was reassuring (Fig. 5). Laparoscopy was performed to reduce the gastric herniation. The stomach was dilated and intact without signs of ischemia. The periesophageal attachments anteriorly were intact. The distal gastric antrum, pylorus, and proximal duodenum were posterior to the esophagus, above the diaphragm extending through a paraesophageal hernia. While a suction catheter was within the esophagus, to ensure a normal lumen, the hiatus was closed posteriorly. A Nissen fundoplication was performed followed by placement of a laparoscopic gastrostomy tube. Radiographs obtained 6 weeks after surgery demonstrated no evidence of recurrent hernia (Fig. 6).

The patient later underwent genetic testing which was positive for mutation in the HDAC8 gene on the X chromosome, diagnostic of Cornelia de Lange Type V. The family underwent genetic counseling. The mutation was found to be de novo and not detected in either parents' DNA sample. The importance of multidisciplinary surveillance was explained.

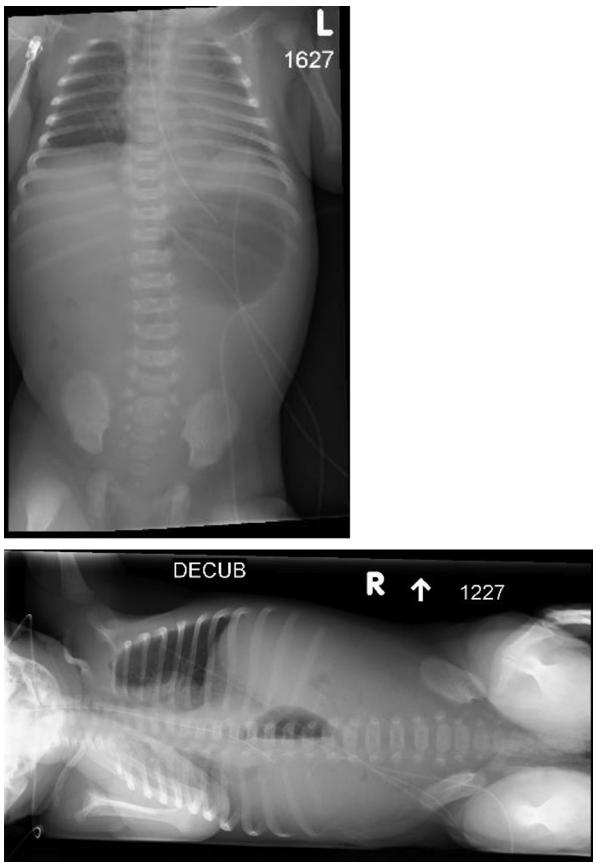

Fig. 3 – Day of life 7. Increased gastric distension with a spherical contour in the left upper quadrant. There is a paucity of additional bowel gas. The enteric tube takes the expected course and the tip is seen within the stomach, below the left hemidiaphragm. There is a fluid level in the dilated lucency in the left upper quadrant, thought to be the stomach, on the decubitus film.

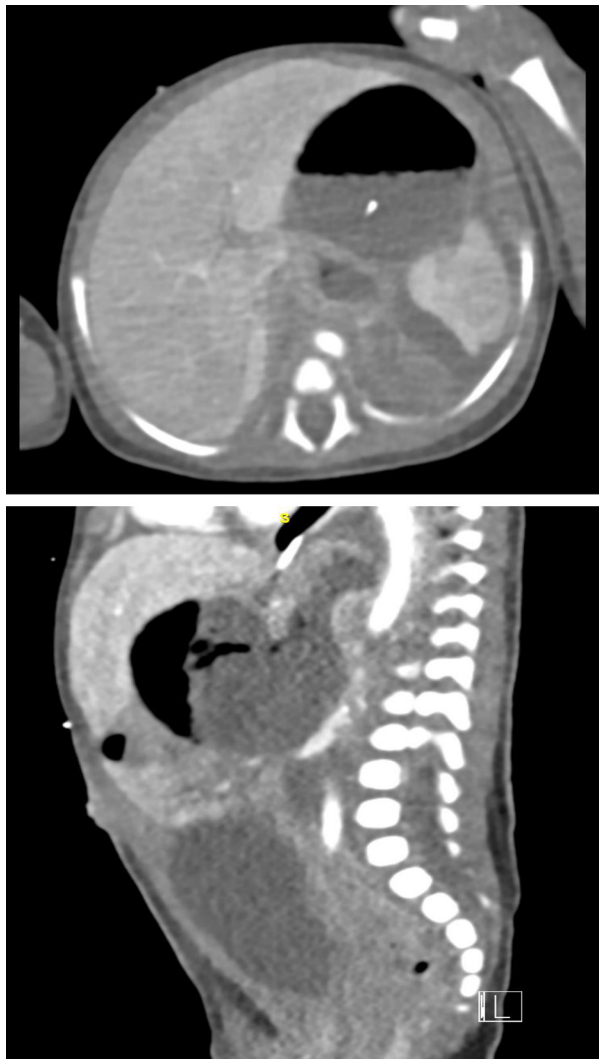

Fig. 4 – Day of life 7. Axial CT demonstrates the feeding tube passing through a normally positioned gastroesophageal junction into the gastric lumen, anterior to a herniated pylorus. Sagittal CT demonstrates herniation of the gastric antrum above the diaphragm, posterior to the gastroesophageal junction.

Discussion

Cornelia de Lange Syndrome (CdLS) is a rare multisystem disorder with variable phenotypes depending on the genes affected [1]. In our case, the patient's mutation was X-linked explaining the variable clinical manifestations that can be seen between different patients. Several typical characteristics include dysmorphic facies, developmental delay, growth retardation, neurologic problems, limb abnormalities, and gastrointestinal manifestations such as GERD and congenital diaphragmatic hernia (CDH) [2]. CdLS is thought to affect up to 1 in every 10,000-30,000 births [2].

There is a known associated association with CDH with a wide reported variance, thought to affect up to 5%-20% of patients with CdLS [2]. For comparison, CDH is estimated to occur 1 in every 2000-3000 live and still births overall [3]. In a study of 13 patients with CdLS, 1 patient had a congenital diaphragmatic hernia [3]. In a registry of 9251 patients with congenital diaphragmatic hernia, Gupta et. al identified 21 patients who had both CDH and CdLS [2]. Further classification of the type of congenital hiatal hernias were not specified.

Most congenital diaphragmatic hernias described in the pediatric literature are Bochdalek and Morgagni type with a small percentage being congenital hiatal hernias, with hiatal hernias mostly described in case reports [4,5]. A majority of hiatal hernias in pediatric patients are acquired [4]. A retrospective review of congenital paraesophageal hiatal hernias by Youseff et. al over a period from 1988 to 2013 demonstrated 2 of 14 patients with congenital type IV hiatal hernia, or paraesophageal hernia [5]. In their small sample size, the most common symptom in type IV hernias was respiratory distress [5].

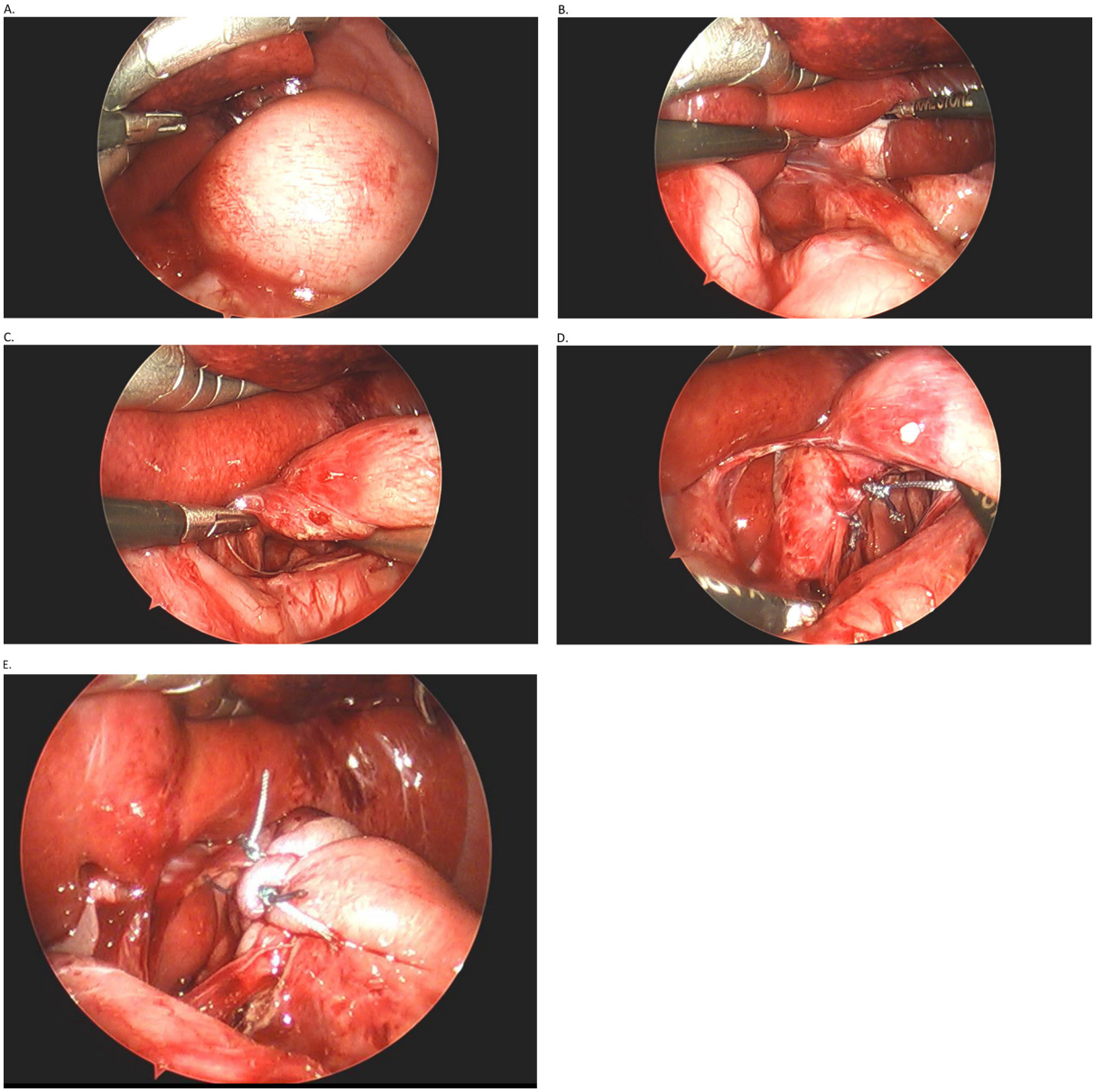

Fig. 5 – Gastric distension was noted despite orogastric decompression. (B) The esophageal ligaments were in place anteriorly. (C) The hiatal defect is noted after reduction of the contents. (D) Hiatal repair. (E) Nissen fundoplication.

Our case presents not only an uncommon genetic mutation but also an association with a rare type of congenital hiatal hernia. We are not aware of a reported type IV congenital hiatal hernia, or paraesophageal hernia, presenting in a patient with Cornelia de Lange syndrome. Though the disease has a known rare association with diaphragmatic hernias, there is a wide phenotypic presentation and limited literature further describing the hernias present in the small cohort of patients who have both CdLS and congenital diaphragmatic hernias.

This case was challenging for several reasons. The patient had additional medical issues that may have delayed complete characterization of the mediastinal contour abnormality, which was eventually recognized as a type IV hiatal hernia, or paraesophageal hernia. Secondly the patient was NPO for the first several days of life secondary to her neurologic status, which may have further delayed diagnosis. Additionally, the patient was not evaluated with an upper GI exam or oral contrast CT to demonstrate the degree of physiologic obstruction, although gastric obstruction was suspected due to emesis and gas distended stomach on exam and during surgery.

Though rare, congenital paraesophageal hernia is an important diagnosis to entertain in the newborn with gastrointestinal complaints and abnormal contour on chest radiograph. Consideration of underlying genetic mutation, espe-

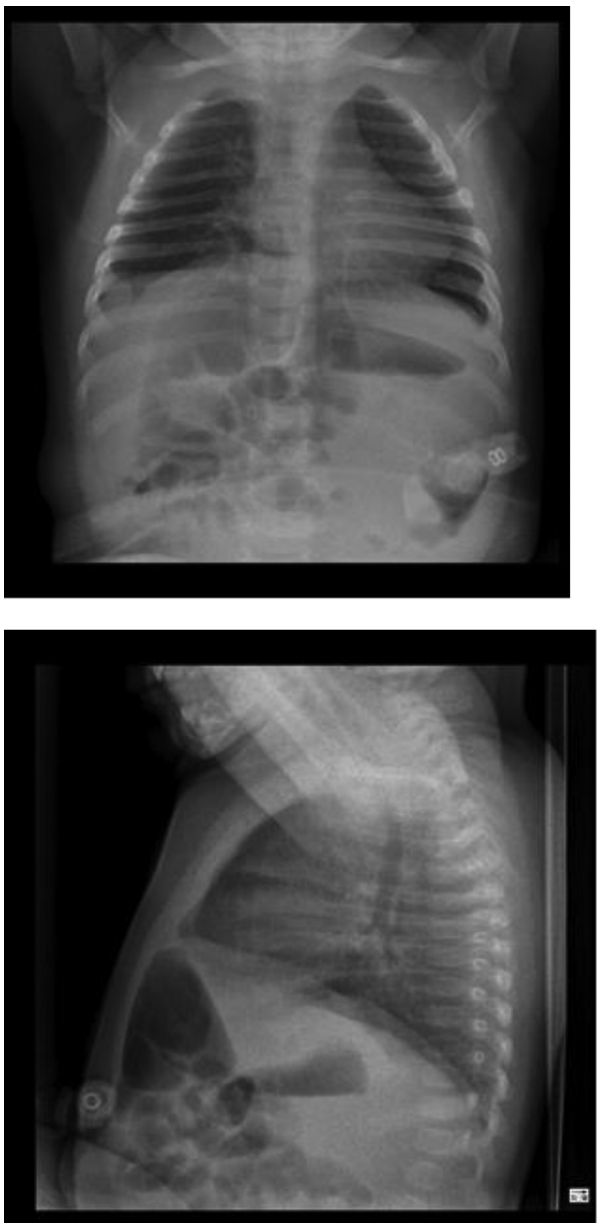

Fig. 6 – Chest radiograph notes intact repair at 6 weeks postop.

cially if physical exam demonstrates abnormalities, should be considered.

Conclusion

Cornelia de Lange Syndrome is a rare syndrome with multisystemic effects and a wide phenotypic spectrum. In patients with known or suspected Cornelia de Lange Syndrome, it is important to be aware of the symptoms, imaging findings, and treatment of congenital diaphragmatic hernias, including the rarer types of paraesophageal hernias. In patients with suspected or confirmed congenital diaphragmatic hernias, it is also important to entertain the possibility of a genetic syndrome.

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