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Emesis in a Term Infant with Dysmorphic Features

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PRESENTATION

A female infant is born at 38 weeks' estimated gestational age to a 23-year-old, gravida 2, para 2 woman. Maternal history is significant for poorly controlled chronic hypertension and limited prenatal care. The woman reports she was returning home from vacation when she experienced painful, frequent contractions, prompting presentation to the nearest emergency department. Her initial blood pressure is 152/102 mm Hg. She has spontaneous rupture of membranes and undergoes repeat cesarean section 3 hours after arrival at the academic medical center.

In the delivery room, Apgar scores are 7 at 1 minute (0 for skin color, and 1 for respiratory effort) and 9 at 5 minutes (1 for skin color). Growth parameters are consistent with a small-for-gestational age infant; the newborn's head circumference, weight, and length are all less than the 1st percentile for gestational age. Initial physical examination is remarkable for upper limb abnormalities, including oligodactyly with the presence of only 2 formed digits on the left upper extremity, a single palpable forearm bone in the left upper extremity, a sacral dimple, significant hirsutism, and abnormal facial features, including synophrys and highly arched eyebrows. She is admitted to the NICU for further care.

In the NICU, she is initiated on feeds of expressed breast milk and colostrum. She passes meconium on the first day after birth. The neonate initially tolerates small volumes of feeds, but by day 2, she experiences nonbilious emesis 15 to 30 minutes after each feed. Intravenous fluids are initiated and a Replogle tube is placed. An abdominal radiograph is obtained on the second day after birth (Fig 1).

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DIFFERENTIAL DIAGNOSIS

The differential diagnosis for recurrent emesis and feeding intolerance in this infant with a nonobstructive bowel gas pattern on the abdominal radiograph includes:

- Gastroesophageal reflux
- Vascular ring or sling
- Duodenal web
- Duodenal stenosis
- Annular pancreas
- Malrotation with or without midgut volvulus
- Extrinsic compression
- Necrotizing enterocolitis

CASE PROGRESSION

The infant continues to have persistent emesis with all feeds. On day 4 after birth, she undergoes a fluoroscopy upper gastrointestinal (GI) series (Video 1, Fig 2). She continues to vomit regularly with feeds, prompting advancement to a nasojejun tube for enteral nutrition. Her feeding tolerance improves. During this interval, a genetics consultation is obtained, and there is high suspicion for Cornelia de Lange syndrome (CdLS), given the infant's physical examination findings.

On day 11 after birth, the infant develops bilious emesis, prompting surgical consultation; a fluoroscopy upper GI series is repeated (Video 2, Fig 3), which uncovers no evidence of volvulus, but confirms her previously known malrotation. In addition, the study demonstrates an abnormally dilated duodenal bulb with abrupt transition at the D1/D2 junction, concerning for a Ladd band causing intermittent obstruction in this infant.

The infant continues to have projectile emesis, and on day 12 after birth, she is taken to the operating room for Ladd procedure and insertion of a gastrostomy feeding tube. Operative notes report "classic features of intestinal malrotation without volvulus, with significant Ladd bands entrapping the duodenum in the right upper quadrant and retroperitoneum requiring Kocherization of the duodenum and division of Ladd bands."

Postoperatively, she tolerates feedings via her gastrostomy tube without difficulty. She remains in the NICU for advancement of feeds and coordination of care before discharge and travel back to the family's home state. The infant is discharged from the NICU 29 days after birth, with close follow-up arranged with her local pediatrician.

Final Diagnosis

The infant is diagnosed as having intestinal malrotation with duodenal obstruction secondary to Ladd bands.

DISCUSSION

CdLS is a well-known syndrome associated with multiple congenital anomalies (cardiac, GI, renal, genital, and ocular), distinct facial features, limb abnormalities, growth restriction, and developmental delay. (1) A recent consensus statement published by international experts highlights the molecular and clinical features for CdLS in both classic and nonclassic phenotypes. (2) Feeding difficulties and failure to thrive are commonly observed in this patient population. (3)

GI abnormalities are a frequent cause of morbidity and mortality in CdLS, with reflux being the most prevalent GI manifestation. (4) Other associated GI system pathologies include pyloric stenosis, imperforate anus, malrotation with or without volvulus, congenital diaphragmatic hernia, and small-bowel duplication. (2) Although these diseases may vary in their presentation, many are potentially deadly. One study identified the GI system as the second most common cause of mortality in CdLS behind respiratory pathology; specifically, GI-related deaths were most often due to volvulus or obstruction. (5)

For the current patient, intestinal malrotation presented on day 4 after birth. Malrotation is seen with a higher frequency among patients with CdLS in comparison to the general population; the incidence of symptomatic malrotation was reported to be 5% to 10% in two separate series of patients with CdLS. (6)(7) This incidence is significantly higher than age-matched controls, in which symptomatic malrotation is seen in only 1 in 6,000 live births. (8)

Clinically, our patient initially had projectile, nonbilious emesis, which progressed to bilious emesis on day 11 after birth. This clinical change is attributable to the development of duodenal obstruction from Ladd bands. Fortunately, malrotation in our patient is not complicated by volvulus with small-bowel ischemia, but patients with malrotation are certainly at risk to develop this life-threatening complication. In many cases, malrotation initially presents as one of these acute complications; notably, acute cecal volvulus is the most common presentation of malrotation in patients with CdLS. (5)(6)

As evidenced before, neonates with CdLS are at increased risk of having malrotation, which predisposes them to possible complications such as volvulus or duodenal obstruction. As such, clinicians must have a high index of suspicion for malrotation in any infant with CdLS who develops feeding difficulties, emesis, abdominal distention, or change in behavior or mental status. Appropriate clinical management is prudent in such scenarios, because these infants are at increased risk for morbidity and mortality if not properly identified in a timely manner.

Lessons for the Clinician

- CdLS is a multisystemic syndrome associated with multiple congenital anomalies (cardiac, GI, renal, genital and ocular), distinct facial features, limb abnormalities, growth restriction, and developmental delay. (1)
- Malrotation is a clinically significant finding in CdLS and should be considered in these patients if they present with feeding intolerance.

References

1. Deardorff MA, Noon SE, Krantz ID. Cornelia de Lange syndrome. GeneReviews. Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1104/>. Accessed August 6, 2019
2. Kline AD, Moss JF, Selicorni A, et al. Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. *Nat Rev Genet*. 2018;19(10):649–666
3. Bull MJ, Fitzgerald JF, Heifetz SA, Brei TJ. Gastrointestinal abnormalities: a significant cause of feeding difficulties and failure to thrive in Brachmannde Lange syndrome. *Am J Med Genet*. 1993;47(7):1029–1034
4. Luzzani S, Macchini F, Valadè A, Milani D, Selicorni A. Gastroesophageal reflux and Cornelia de Lange syndrome: typical and atypical symptoms. *Am J Med Genet A*. 2003;119A(3):283–287
5. Schrier SA, Sherer I, Deardorff MA, et al. Causes of death and autopsy findings in a large study cohort of individuals with Cornelia de Lange syndrome and review of the literature. *Am J Med Genet A*. 2011;155A(12):3007–3024
6. Mariani M, Decimi V, Bettini LR, et al. Adolescents and adults affected by Cornelia de Lange syndrome: A report of 73 Italian patients. *Am J Med Genet C Semin Med Genet*. 2016;172(2):206–213
7. Kline AD, Grados M, Sponseller P, et al. Natural history of aging in Cornelia de Lange syndrome. *Am J Med Genet C Semin Med Genet*. 2007;145C(3):248–260
8. Aboagye J, Goldstein SD, Salazar JH, et al. Age at presentation of common pediatric surgical conditions: Reexamining dogma. *J Pediatr Surg*. 2014;49(6):995–999 [PubMed: 24888850]

American Board of Pediatrics Neonatal-Perinatal Content Specifications

- Know the clinical features and inheritance patterns of common syndromes or associations that can be recognized in the newborn period (eg, VATER association and DiGeorge syndrome).
- Know the etiology, clinical manifestations, diagnosis, and management of GI strictures.

Figure 1.
The abdominal radiograph obtained on day 2 after birth demonstrates a non-obstructive bowel gas pattern but with asymmetrical distension of bowel in the left abdomen.

Figure 2. Spot image from the upper gastrointestinal series obtained on day 4 after birth showing the abnormally positioned duodenal sweep far anterior to the spine (arrow).

Figure 3. Spot image from the repeat upper gastrointestinal series showing a severely dilated duodenal bulb (upper arrow) and a small amount of contrast moving into the distal duodenum along the feeding tube (lower arrow).

Video 1.

Upper gastrointestinal series obtained on day 4 demonstrates an anteriorly positioned duodenal sweep and intermittent dilation of the duodenal bulb without fixed narrowing. No evidence of midgut volvulus is seen. Although there is intermittent dilation of the duodenal bulb, there is no evidence of fixed narrowing to suggest duodenal stenosis or extrinsic compression.

Video 2.

Repeat upper gastrointestinal series 7 days after the initial study, demonstrating severe persistent dilation of the duodenal bulb but with slow passage of contrast through the duodenal sweep into the jejunum, following the course of the nasojunal feeding tube. Note the inferior displacement of the ligament of Trietz with the tube in place in this infant with known malrotation.