

Index of Suspicion in the Nursery

1 A Full-Term Neonate with Trisomy 13 and Pneumoperitoneum

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CASE PRESENTATION

A female infant is born at 37+1 weeks of gestation via an emergency cesarean section to a 35-year-old, gravida 3, para 1+1 woman who is taking insulin for a known medical history of gestational diabetes mellitus but has no other medical problems. The pregnancy is normal, with normal prenatal laboratory screening results; a fetal survey is remarkable, because it reveals Dandy-Walker malformation. There is no history of consanguinity. The mother presents with labor pain and does not receive antenatal corticosteroid therapy.

The neonate's Apgar scores are 9 and 9 at 1 and 5 minutes, respectively. Her vital signs are stable (temperature, 36.5°C; heart rate, 160 beats/min; respiratory rate, 75 breaths/min; blood pressure, 60/35 mm Hg; and oxygen saturation, 90%), though initial newborn examination reveals dysmorphic features (depressed nasal bridge, large globular nose, polydactyly in 1 hand and both feet, thin upper lip, and sacral agenesis).

The infant is born with a birthweight of 2,400 g (2nd percentile), length of 48 cm (27th percentile), and head circumference of 32 cm (6th percentile).

The infant immediately develops respiratory distress and desaturation, requiring a transfer to the NICU with a continuous positive airway pressure of 5 cm H₂O. The fraction of inspired oxygen (FiO₂) is 23. A partial sepsis evaluation is initiated, and the infant is started on ampicillin and gentamicin. Respiratory distress improves within a few hours; her blood culture yields no organisms, and antibiotics are discontinued within 48 hours. Immediate genetic consultation is requested.

Karyotyping confirms the diagnosis of trisomy 13 "(47,XX, +13 [free in all cells])." Magnetic resonance imaging confirms the diagnosis of Dandy-Walker malformation.

The neonate starts formula feeding on day 2 after birth. She tolerates the feedings until day 4, when she develops vomiting and abdominal distention. Abdominal radiography shows pneumoperitoneum (Fig).

DISCUSSION

Differential Diagnosis

The differential diagnosis for abdominal distention and pneumoperitoneum in a newborn with trisomy 13 includes the following:

- Necrotizing enterocolitis
- Intestinal malrotation with perforation
- Sepsis
- Intestinal perforation

AUTHOR DISCLOSURE Drs Alallah and Al Talhi have disclosed no financial relationships relevant to this article. This report does not contain a discussion of an unapproved/investigative use of a commercial product/device.

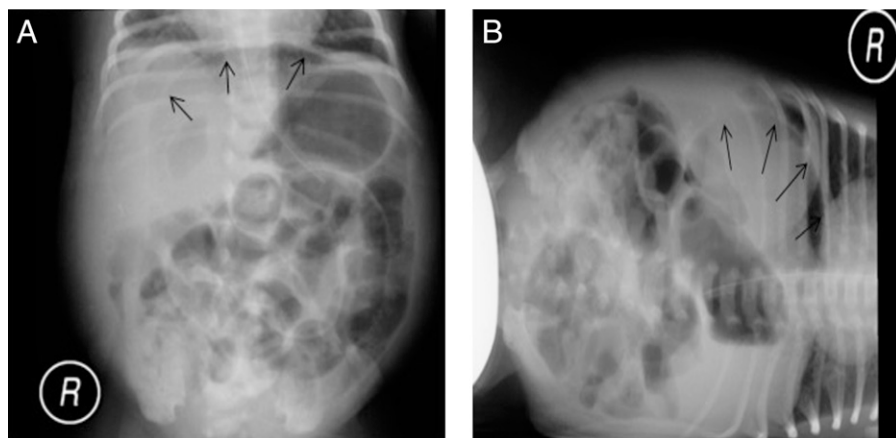


Figure. Evidence of pneumoperitoneum in both the supine (A) and left decubitus (B) positions (black arrows).

Actual Diagnosis

Intestinal perforation arising from spontaneous perforation of Meckel diverticulum.

Case Progression

The infant was given nothing by mouth (*nil per os* [NPO]) and was started on treatment with 3 antibiotics: vancomycin, gentamycin, and metronidazole. Blood culture was positive for *Klebsiella pneumoniae*. She was taken to the operating room for a laparotomy where a perforated Meckel diverticulum was found 10 cm from the ileocecal valve. Resection of the diverticulum and the appendix with end-to-end anastomosis of the bowel was performed. She came back to the NICU after surgery in a good, stable condition. She was kept NPO and continued to receive antibiotics until an improvement was noted. Feeding started after a few days, which she tolerated until full feeds. She was discharged from the hospital in a stable condition at 23 days of age.

Before discharge from the NICU, she was assigned a code (“do not resuscitate”), because of her trisomy 13 with brain malformation.

After 45 days, she presented with a cough, fever, cyanosis, and apnea of 2 days’ duration, as well as a history of contact with a patient with an upper respiratory infection secondary to influenza A (H1N1) virus infection. A diagnosis of viral pneumonia was confirmed for type A influenza, with a negative blood culture. Three months later, she presented to the emergency department with a fever, productive cough, and decreased activity and feeding of 2 days’ duration. She appeared toxic, with mild respiratory distress, and was diagnosed with respiratory syncytial virus pneumonia and hyponatremic dehydration. During the course of hospital admission, her status deteriorated and she eventually died.

THE CONDITION

Trisomy 13 (Patau syndrome) is an extremely rare, yet serious aneuploid problem that occurs in 1 in 12,000 births, with a 91.4% mortality rate within the first year after birth. It results from either nondysjunction or Robertsonian translocation. The observed recurrence rate is 1%, albeit with the possibility of increasing because of maternal age and the chromosomal analysis result. Trisomy 13 involves multiple apparent malformations and facial dysmorphisms from the head to the extremities, aplasia cutis congenita being one of its pathognomonic characteristics. The most common malformations are median facial or central nervous system defects; the majority of those with the condition suffer from congenital heart defects. Meckel diverticulum, an unrelated issue, is a rare misconfiguration of the small intestine that occurs in 1% to 2% of those with trisomy 13 worldwide. (1) The coexistence of trisomy 13 and Meckel diverticulum in the same patient is unreported, to the best of our knowledge, aside from scattered case reports of trisomy 13 autopsies. Here, we report a case of a full-term neonate with both trisomy 13 and Meckel diverticulum who presented with abdominal distention and perforation.

The existence of perforated Meckel diverticulum in a newborn without trisomy 13, however, has been reported. (2) Colacino and Petterson described a postmortem examination of 4 cases of which 1 had Meckel diverticulum. (3) Petterson found Meckel diverticulum on autopsy in a 6-year-old boy with trisomy 13 (4); Aziz also reported multiple anatomic anomalies in a trisomy 13 case in which Meckel diverticulum was noted. (5) Finally, in a US study of the profiles of some patients with trisomy 13, 3 patients had Meckel diverticulum. (6)

More related reports were made about 10 years ago—Balci et al reported Meckel diverticulum in a preterm neonate

with trisomy 13 after dissection (7) and Iijima et al discovered Meckel diverticulum by accident in a preterm with trisomy 13 while performing laparotomy for an omphalocele. (8)

In summary, Meckel diverticulum in newborns with trisomy 13 has only been discovered on autopsy while managing another disease, or as in this case, managing abdominal distention with perforation.

The exact magnitude of Meckel diverticulum among newborns with trisomy 13 is not yet determined.

Lessons for the Clinician

- Abdominal distention and pneumoperitoneum in a newborn with trisomy 13 should alert neonatologists to check for congenital malformations, especially of the gastrointestinal tract.
- The coexistence of Meckel diverticulum with trisomy 13 should be considered a possibility.

American Board of Pediatrics Neonatal-Perinatal Content Specifications

- Know the morphogenesis of the gastrointestinal tract and the congenital malformations of the gastrointestinal tract.

- Know the differential diagnosis of abdominal distention and perforation in the neonate.
- Know the association between trisomy 13 and gastrointestinal-tract malformations.

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