T
disomy 13 (T13) or Patau's syndrome is an extremely rare but serious aneuploidy that occurs in 1 in 12,000 births, with an 86–91% mortality rate within the first year after birth. It results from either nondisjunction or Robertsonian translocation.\textsuperscript{1} The most common malformations are median facial or central nervous system defects, and the majority of those with T13 also suffer from congenital heart defects.\textsuperscript{1} About 25% of T13 patients are also affected by another genetic condition, Meckel's diverticulum (MD), which almost always remains asymptomatic. Clinical presentation of MD in T13 neonates is extremely rare.\textsuperscript{2,3} Only two symptomatic cases of infants with T13 and MD are described in the literature. Here, we report a case of a full-term neonate with T13 and MD who presented with bilious aspirates and intermittent volvulus due to MD with band.

**CASE REPORT**

A full-term male infant (3671 g, 41 + 2 weeks gestation), was delivered to a 27-year-old primagravida by normal vaginal delivery. The neonate had Apgar scores of 6 and 8 at 1 and 5 minutes, respectively. The pregnancy had been normal, with normal prenatal serological results and normal antenatal ultrasound results in the second trimester. The parents were second-degree consanguineous.

On physical examination, the baby was noticed to have multiple congenital anomalies including complete bilateral cleft lip/palate, low-set ears, microphallus, and bilateral undescended testes. Further evaluation after admission to neonatal intensive care unit revealed more anomalies. Cranial ultrasound revealed vermian hypoplasia and cystic malformation of the posterior fossa. An echocardiography showed moderate patent ductus arteriosus (PDA), small atrial septal defect (ASD), and bicuspid aortic valve. The newborn was intubated and mechanically ventilated to mitigate respiratory distress. The abdomen felt soft on palpation and appeared not-distended, with normal passage of meconium. Feeds were initiated at 5 mL every three hours on day two of life. Feeds were tolerated well for two days but on the third day, greenish aspirates were noted. The abdomen remained soft and non-distended. An upper gastrointestinal (GI) contrast study showed the duodenaljejunal flexure to be in a normal position, but the upper bowel loops had a corkscrew appearance raising concern for volvulus (Figures 1 and 2).

Upon exploratory laparotomy, MD was found with a peritoneal band extending from base
of diverticulum to umbilicus, possibly causing intermittent volvulus and the consequent intestinal obstruction. A small cyst-like structure was noted in the middle of the band. No caliber change of bowel was noted at that level. The MD was resected, and the bowel underwent primary anastomosis. Feeds were started on the fourth postoperative day and increased quickly to full feeds. Karyotyping revealed T13, 47, XY + 13. The child kept tolerating feeds and went on to have an uneventful laparoscopy-assisted gastrostomy insertion. Pathology of the resected segment confirmed vitello-intestinal remnant on a small bowel segment in keeping with intraoperative findings. When correlated with the history of T13 and the clinical presentation, the contrast examination findings led us to conclude that the bowel obstruction was most likely due to symptomatic MD.

**DISCUSSION**

T13 is one of the most common autosomal trisomies, accompanied by anomalies in several organs. The relation of T13 to a clinical syndrome was first recognized by Patau et al, in 1960 and it is also known as Patau’s syndrome. Median survival time for patients with T13 is only 7–10 days. Between 86% and 91% of live-born babies with T13 do not survive beyond the first year of life. Survival beyond the first year has been associated with mosaicism. A 19-year-old girl is the oldest known living person with regular T13.

MD results from the formation of a true diverticulum in the small intestine as a result of an incomplete obliteration of the vitelline duct. It is often defined informally by the rule of twos: (1) occurs in approximately 2% of the population with a male-to-female ratio of 2:1, (2) is located within two feet from the ileocecal valve, (3) may manifest before the age of two years, and (4) is 2 inch long (though in practice variable). Although MD is a common congenital anomaly of the gastrointestinal tract, its symptomatic manifestation in the neonatal period is rare. Common presentations of neonatal MD that have been reported in the literature include bowel obstruction by inflammation, perforation, intussusception, segmental ileal dilatation, and ileal volvulus.

The coexistence of T13 and symptomatic MD has been reported in only two cases based on our literature search, although there are scattered reports of such coexistence seen during autopsies.
of T13 patients. In this case, MD was discovered at the time of laparotomy for suspected volvulus with feed intolerance and bilious aspirates. MD has been diagnosed and reported earlier both during exploratory laparotomy for suspected volvulus/malrotation and more so as an incidental finding in a postmortem of T13 patients.6,13–18

Considering the rarity of symptomatic presentation of MD in T13 patients, it is not cost-appropriate to routinely recommend an isotope scan except perhaps in presence of suggestive GI symptoms. We think that even if the isotope scan (known to have a 30% false negative rate) is negative in a baby with features of volvulus/malrotation, there is still the need for urgent surgical exploration and correction.

In our case, a significant point of learning is reflected in the review of the upper GI contrast examination. The radiologist performing the upper GI contrast study should look for the proximal small bowel loops for their location, morphology, and contrast emptying, even though a clear demonstration of the normal duodenojejunal flexure is made. Otherwise, volvulus without malrotation can be missed.

The approach to the treatment of MD depends on whether it is discovered incidentally or as a result of symptoms. A physician may choose whether or not to surgically resect an incidentally discovered asymptomatic MD. However, a case of symptomatic MD must be treated by surgical resection, as in the current case.

CONCLUSION

Preoperative diagnosis of a symptomatic MD in a neonate with T13 is difficult, so it is necessary to maintain a high index of suspicion aided by delayed images of the contrast study. An early surgical intervention is recommended for a successful outcome. The possibility of MD should be considered in any child with T13 that presents with bowel obstruction, especially intermittent. Standard surgical treatment comprising MD resection and primary bowel anastomosis is generally safe and effective.

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Disclosure

The authors declared no conflicts of interest. Consent was obtained from the patient’s parents.