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Ileal atresia, malrotation and Hirschsprung's disease: A case report

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ABSTRACT

Ileal atresia associated with malrotation is rare, but the additional diagnosis of Hirschsprung's disease (HD) in the same patient has not been described in the literature. This case report presents a newborn with a bowel obstruction requiring surgery for ileal atresia and malrotation. The post-operative course was complicated by a distal bowel obstruction. Evaluation of the resected bowel from the original surgery was consistent with total colonic aganglionosis, and a diverting ileostomy was performed. Delay in diagnosis of HD is not uncommon when an initial gastrointestinal anomaly is diagnosed and should be suspected when bowel function fails to return after corrective surgery.

Atresia of the intestinal tract associated with malrotation is rare, but the additional diagnosis of Hirschsprung's disease (HD), or total colonic aganglionosis, in the same patient does not appear in the literature. Jejunoileal atresia (JIA) occurs in approximately 1 in 5000 live births and occurs equally in males and females [1–4]. In rare instances, JIA has been found to be associated with HD, malrotation and other disorders [1,5–7]. The coexistence of these conditions can complicate the evaluation of bowel obstruction, and the co-diagnosis of HD is often not confirmed appropriately at the time of laparotomy. This article presents an infant who had ileal atresia with malrotation further complicated by total colonic aganglionosis. We report a unique combination of gastrointestinal anomalies in the same patient that has not been previously reported.

1. Case report

A full-term male infant was born via an uncomplicated spontaneous vaginal delivery with a birth weight of 3.4 kg in a community hospital. He developed progressive abdominal distention during the first day of life and did not pass meconium. After 2 bouts of bilious emesis, transfer was arranged for further evaluation and management in a tertiary medical center. On admission, the patient was in mild distress. His abdomen was distended, and bowel sounds were present. The anus was patent with normal sphincter tone, and a small amount of meconium was present on rectal stimulation. No other malformations were apparent on physical exam. Abdominal roentgenograms (AXR) demonstrated numerous loops of bowel in a right-sided abdominal distribution and a separate enlarged loop in the left lower quadrant. No free air was appreciated. As well, on the plain film, hemivertebral bodies were seen at the distal thoracic spine with dysplasia of the left ribs. Concern for a distal bowel obstruction was evaluated by a contrast enema that showed a decreased caliber, but not micro colon, located in the left side of the abdomen. An upper gastrointestinal contrast study revealed passage of contrast in a corkscrew fashion in the right upper quadrant without crossing midline consistent with malrotation, (Fig. 1). Exploratory celiotomy revealed a type IIIb ileal atresia as well as malrotation with a narrow mesenteric base and no volvulus. The cecum and 5 cm of atretic terminal ileum were in the left lower quadrant (Fig. 2). A Ladd procedure and ileocecectomy followed by ileoascending colostomy were performed. Post-operatively, the patient recovered well in the neonatal intensive care unit and received total parental nutrition while awaiting return of bowel function. After 2 weeks, he tolerated complete nutritional support orally and passed smears of meconium, however only with rectal stimulation. In the third post-operative week he developed abdominal distension and obstipation. An AXR revealed dilated loops of bowel without signs of perforation or pneumatosis. A repeat contrast enema on post-operative day 24 revealed a patent anastomosis with no sign of distal obstruction. The next day, a repeat AXR showed persistent dilated bowel loops

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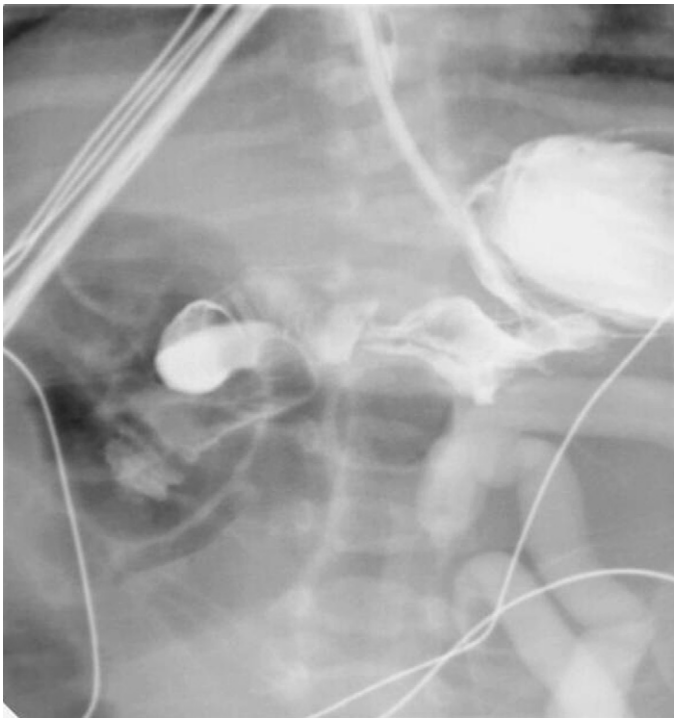

Fig. 1. Upper gastrointestinal study showing contrast with corkscrew path into right upper quadrant without crossing midline. As well, residual contrast from water soluble enema showing decreased caliber colon, exclusively left-sided.

with residual contrast in the colon. The pathologic specimens from the initial surgery were re-reviewed and found to be consistent with total colonic aganglionosis. A diverting ileostomy was performed after confirming the presence of ganglion cells in the distal ileum. Post-operatively, bowel activity returned, and the infant tolerated oral feedings. A high output ileostomy was controlled by diet modification and anti-diarrheal agents. At age 11 months, the infant underwent a Soave endorectal pull-through procedure, including a total abdominal colectomy, ileoanal anastomosis, and diverting ileostomy. His post-operative course was uncomplicated. Eight months later, the ostomy was reversed, and stool frequency was controlled with oral loperamide resulting in an average of 4–5 stools per day. At 2 years of age, he was in the 40th percentile for weight and remained on the growth curve.

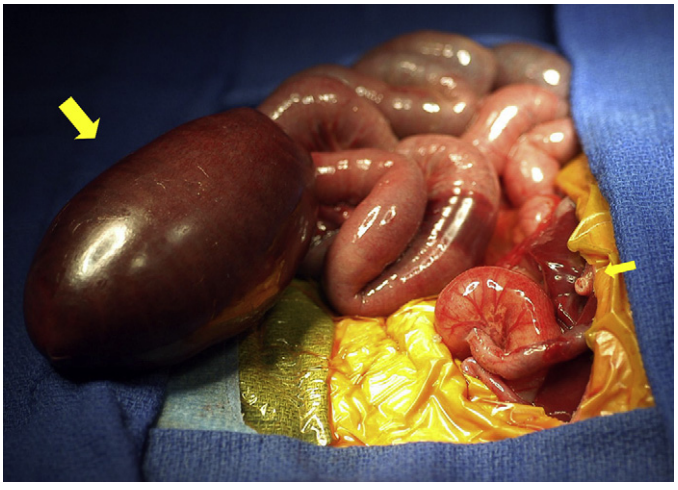

Fig. 2. Distal ileal atresia (Type IIIb) with the proximal bowel terminated in a bulbous blind end (large arrow) and a large mesenteric defect. The collapsed terminal ileum and cecum are located on the left side of the abdomen in close proximity to the umbilical vein (small arrow).

2. Discussion

We describe the unique combination of 3 acute intestinal anomalies in a newborn male not previously reported. Initial evaluation for a distal obstruction did not establish a diagnosis, but showed a decreased caliber colon. Upper gastrointestinal series demonstrated malrotation followed by identification of ileal atresia in the operating room. The expected delay in return of normal bowel function associated with the atresia repair and Ladd procedure, as well as the unlikely combination of all 3 abnormalities, contributed to the late diagnosis of HD. Although the presence of HD in patients with colonic atresia has been well documented [8–10], it is less frequently associated with ileal atresia. A literature review revealed 21 reports of ileal atresia associated with HD; the most recent report by Daher et al. described 2 unrelated newborns that presented with distal obstruction and were treated operatively for ileal atresia. One patient suffered early post-operative obstructive symptoms and returned to the operating room. Intraoperative staged biopsies of the terminal ileum and large bowel confirmed the co-occurrence of HD and the newborn was treated initially with a temporary ileostomy. The other patient initially underwent an ileocecectomy; however the pathology specimen evaluation was delayed. Seven days post-operatively the histologic review was consistent with total colonic aganglionosis and, similarly, a temporary ileostomy was performed [11].

Malrotation and HD are relatively common congenital disorders and occasionally occur concurrently. The true incidence of rotational abnormalities of the midgut is difficult to determine, but is estimated to be symptomatic in 1 of 6000 live births [12] and carries a 5–26% risk of intestinal atresia [13]. HD presents in 1 of 5000 newborns and is associated with a variety of other congenital abnormalities including malrotation [14,15]. Filston and Kirks reported 34 cases of malrotation, with an 11% association with Hirschsprung's disease [16]. Fonkalsrud et al. described a case of a newborn that on the first day of life underwent surgical correction of symptomatic intestinal malrotation and midgut volvulus. The hospital course was complicated by the development of obstructive symptoms on post-operative day 10. A work-up revealed the dual diagnosis of intestinal malrotation with HD [17].

3. Conclusion

- If a young patient suffers from failure of bowel function to return in a timely fashion after corrective surgery for malrotation or other congenital gastrointestinal disorder, HD should be considered as a cause for functional distal obstruction and a rectal suction biopsy is warranted.

Informed consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Conflict of interest

None.

Military disclosure

The views expressed in this article are those of the authors and do not reflect the official policy or position of the Department of the Navy, Department of Defense, or the United States Government.

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