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.

Key words: Ileal atresia
Malrotation
Hirschsprung's disease
Total colonic aganglionosis

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 pneumatisos. 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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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 per-
mformed 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 modifi-
cation 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 uncompli-
cated. 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.

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 pro-
dure, 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 obstruc-
tive 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 tempo-
rary ileostomy was performed [11].

Malrotation and HD are relatively common congenital disorders
and occasionally occur concurrently. The true incidence of rota-
tional 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 re-
ported 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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