

CASE REPORT

Ileal atresia and total colonic hirschsprung disease in a 36-week neonate: A case report

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Abstract

Intestinal atresia and hirschsprung disease are two common causes of bowel obstruction in neonates; simultaneous occurrence is rare. This report delineates a 36-week newborn with ileal atresia and total colonic hirschsprung who was referred to our unit due to failure of meconium passage during the first 48 h after birth.

KEYWORDS

aganglionosis, hirschsprung disease, ileum atresia

1 | INTRODUCTION

Intestinal atresia is a significant cause of primary bowel obstruction in neonates, accounting for 22.4% of all cases.¹ On the contrary, hirschsprung disease (HD) is the most frequent congenital intestinal motility disorder.² HD occurs when bowel segments are not supported by neuronal ganglion formation within the myenteric (Auerbach) and submucosal (Meissner) plexus, which promotes gastrointestinal movements.

And in 80% of cases, the absence of ganglion cells is limited to the rectosigmoid area. However, aganglionosis may extend through the colon with the distal ileum (10%) or more proximal segments.³ Also, in these cases, a water-soluble enema shows a “question mark” configuration in the foreshortened colon with no transitional zone.⁴ The rectal biopsy can always confirm the diagnosis.

Concurrent small intestinal tract atresias associated with hirschsprung are <1%.

In this case report, we simultaneously present an infant born with ileal atresia and hirschsprung disease.

2 | CASE PRESENTATION

A 36-week male neonate was referred to the pediatric surgery service on the second day of life because of failure to pass meconium, abdominal distension, and bilious emesis.

The patient was born to a 23-year-old healthy primipara Iranian mother without a history of abortion via normal vaginal delivery. Her birth weight was 2500 g.

On physical examination, the primary vital signs were in the normal range, the bowel sounds were decreased, and the rectum was empty.

Initial abdominopelvic radiography revealed thumb-sized intestinal loops with no gas in the rectum (Figure 1A). Small bowel contrast series confirmed dilated proximal blind end (Figure 1B).

Baseline biochemical and blood gas analyzes were in the normal range. On the third day, with a diagnosis of intestinal obstruction, the patient underwent an exploratory laparotomy after obtaining informed consent from the parent. We identified ileal atresia with a proximal

dilated, blind-ending 1 cm distal bud attached to the ileocecal region (Type IIIa). (Figure 2) The colon appeared as an unused microcolon throughout. After resecting the proximal dilated ileal pouch and ileocecal part, surgery was followed by an end-to-end ileo-ascending colonic anastomosis.

Permanent hematoxylin and eosin (H&E) pathological examination revealed the absence of ganglion cells in the cecum, the appendix, and the rectum and the abundance of ganglia in the ileum (Figure 3).

Due to the persistence of abdominal distention, no defecation on postoperative days, and a histopathologic report that was compatible with total segment HD, the patient was a candidate for re-exploration. (Figure 4).

Our findings were a microcolon and a functional intestinal obstruction at the anastomosis site with no anastomotic leak.

After taking multiple levels of frozen section biopsies to reaffirm the total colonic aganglionosis, the patient underwent an ileostomy and colonic mucus fistula bypass.

The final permanent pathology studies by the calretinin test disclosed total aganglionic colon HD.

Swenson pull-through surgery with removal of all colon and ileoanal anastomosis was performed on a three-month-old. The case tolerated oral feeding after 5 days and was discharged from the hospital in good condition.

3 | DISCUSSION

Coexistence of ileal atresia and total colonic HD is a rare event.^{5,6} Conforming to the currently accepted theories, jejunoileal atresia arises due to intrauterine ischemic vascular events in the third trimester, such as intussusception, perforation, volvulus, or thromboembolism; maternal smoking as a hypercoagulation state^{7,8} and cocaine use as vasoconstrictive medications.⁹

The migration of ganglion cells was completed through the gastrointestinal tract from proximal to distal by 13 weeks post conception. Therefore, early gestational atresia in the sixth to eighth weeks of gestation would result from an ischemic insult to interrupt the caudal migration of ganglion cells and lead to total colonic HD.^{10,11} Finding a very small microcolon, no fibrotic of the left colon, and no meconium distal to atretic segments strengthens this theory.

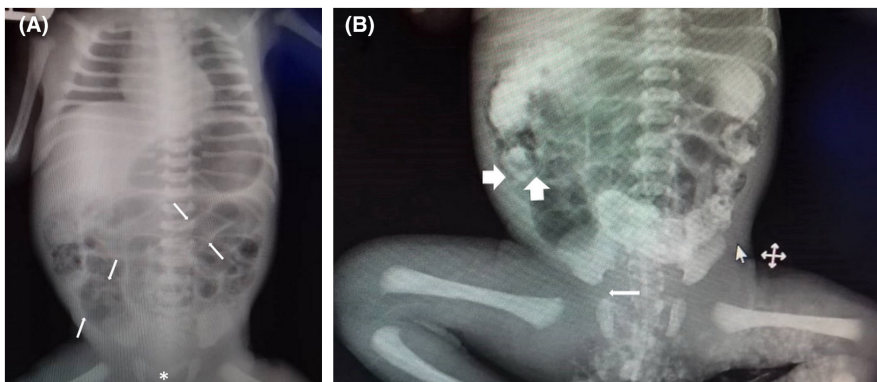


FIGURE 1 (A) several dilated gastrointestinal loops (Arrows) with no gas in the rectum (Asterisk). (B) Small bowel contrast study reveals dilated proximal blind end (Thick arrow).

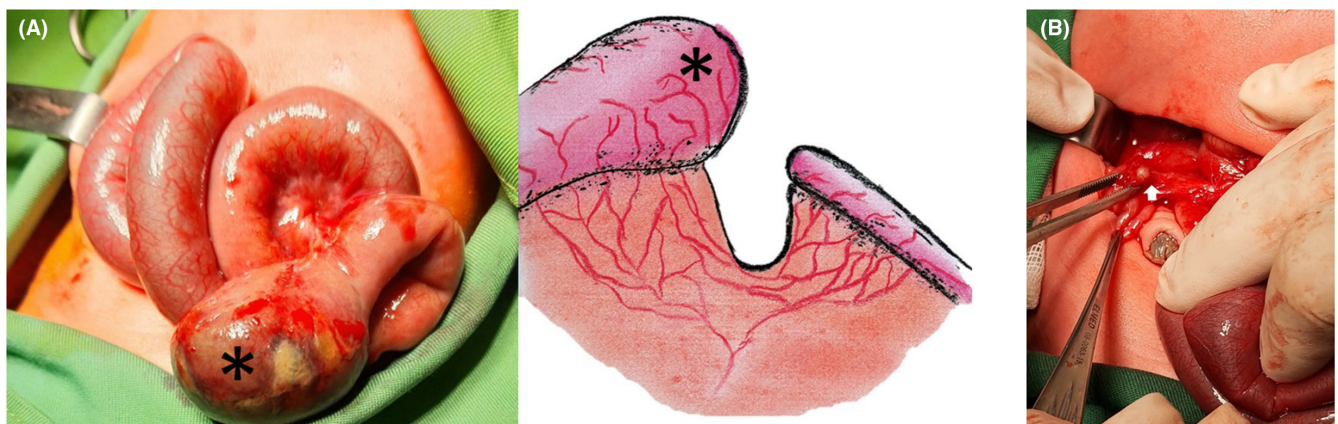


FIGURE 2 (A) Type (III-a) atresia at the laparotomy. The proximal end of the ileum has been marked by the Asterisk. (B) The distal blind end of the ileum is seen as a bud in the ileocecal region (Arrow).

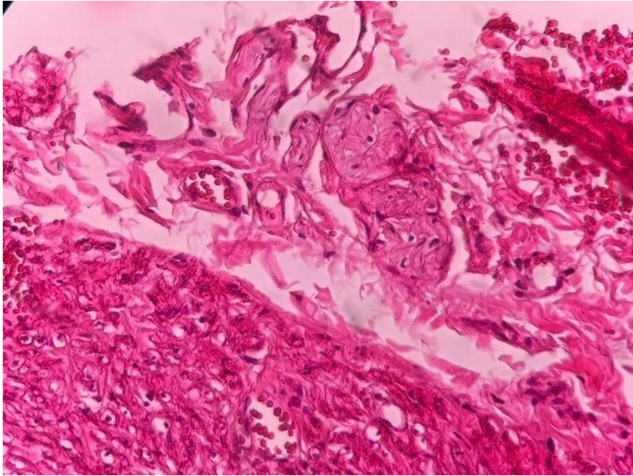


FIGURE 3 The hematoxylin and eosin study of the ileum in the left and colonic specimen in the right revealing the absence of neurologic ganglia compatible with HD ($\times 1000$ magnification).



FIGURE 4 Contrast barium enema demonstrates an unused colon and a functional intestinal obstruction at the anastomosis site.

The other hypothesis that can justify this concurrency is an increased colonic intraluminal pressure and subsequent perforation ileocecal portion due to a developed HD and the secondary small bowel atresia. However, in our case, there was no evidence of meconium spillage into the

peritoneal space during our laparotomy, which weakens the second assumption.

As the common occurrence of microcolon in the cases of distal small intestinal atresia, it is tough to differentiate this colonic appearance during surgery from concomitant total colonic aganglionosis and small bowel atresia.

Therefore, it appears rational to do per-operative colonic biopsies looking for ganglion cells in a frozen section to exclude or confirm the underlying HD in suspicious cases.

A definitive reconstructive operation should be planned once we have established the diagnosis and done a proximal ileostomy.

However, there are controversial questions about the correct timing and the most appropriate treatment options.^{12,13}

Several approaches have been described to treat this, such as primary pull-through without ileostomy or total colectomy with standard techniques (Swenson, Duhamel, or Soave). Neither is superior to the others.

Albeit discriminating, the best operative approach should be constructed based on the surgeon's level of expertise¹³; in our case, we did total colectomy with an ileoanal Swenson procedure when the patient status was allowed.

In conclusion, this rare concurrency should be considered in the cases of small bowel atresia with poor bowel function after the corrective operation.

AUTHOR CONTRIBUTIONS

Khashayar Atqiaee: Supervision; writing – review and editing. **Mehran Hiradfar:** Conceptualization; methodology. **Mehdi Parvizi Mashhadi:** Writing – review and editing. **Ali Samady Khanghah:** Writing – original draft.

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None.

CONFLICT OF INTEREST STATEMENT

The authors declare no competing interests relating to this original work.

DATA AVAILABILITY STATEMENT

The authors declare that they have chosen “Data openly available in a public repository that issues datasets with DOIs” and the data that support the findings of this study will be openly available on the DOI website when prepared.

ETHICAL APPROVAL

We confirm that all named authors have read and approved the manuscript. The protection of the intellectual property associated with this manuscript has been our consideration available on the DOI website when prepared.

CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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