CASE REPORT

Type I sigmoid atresia misdiagnosed as Hirschsprung disease

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Received 3 January 2015; received in revised form 3 February 2015; accepted 26 March 2015
Available online 19 August 2015

KEYWORDS
colonic atresia; contrast enema; Hirschsprung disease; neonatal intestinal obstruction

Abstract Colonic atresia is a rare entity presenting as a neonatal intestinal obstruction. The authors encountered a case of type I sigmoid atresia that was misdiagnosed as Hirschsprung disease in the neonatal period. The case is discussed herein because of the rarity of the condition and to emphasize the importance of a high index of suspicion for this condition.

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1. Introduction

Colonic atresia (CA) is a rare entity with an incidence of one in 20,000 live births and accounts for approximately 1.8–15% of intestinal atresia.1 Neonates with CA typically present with predictably marked and progressive abdominal distension within 24–48 hours after birth. The neonate typically passes little or no meconium.2 Hirschsprung disease (HD), a congenital intestinal neuropathy characterized by the absence of ganglion cells extending from the rectum proximally for a variable distance, presents with the failure to pass meconium within 48 hours after birth and often progresses to abdominal distension and vomiting.3 We report a case of type I sigmoid atresia misdiagnosed as HD in the neonatal period. Diagnosis was missed even on a contrast enema in the postoperative period. The atresia (web) was later diagnosed when colonoscopy was performed to relieve the suspected fecal impaction after colectomy closure. Such complications emphasize the importance of a high index of suspicion for this condition.

2. Case Report

A 5-day-old male child, weighing 2.5 kg, was admitted in the emergency department with complaints of an inability to pass meconium, severe abdominal distension, and bilious vomiting. The child had passed meconium once within 24
hours of life. On examination, the heart rate was 140 beats/min, respiratory rate was 48 breaths/min, and the capillary refill time was < 3 seconds with a severely distended abdomen and palpable bowel loops. Hemogram and blood chemistry were within normal limits, and an X-ray of the abdomen revealed multiple air fluid levels suggesting distal intestinal obstruction. After informed consent was obtained, the patient was prepared for exploratory laparotomy with a right supra-umbilical transverse incision, which revealed a 3-cm-long narrow segment of the rectum proximal to the peritoneal reflection, followed by a zone of transition and a proximal largely distended intestine. A provisional diagnosis of HD was made. Seromuscular biopsy was obtained from the collapsed rectum, and right transverse colostomy was performed. Postoperative recovery was uneventful and the patient was discharged on Postoperative Day 8, with advice to follow-up in the pediatric surgery outpatient department. The patient was lost to follow-up and again attended the outpatient department 6 years after the previous procedure. Barium enema study was performed, which was normal except for a slight narrowing at the sigmoid colon level (Figure 1). A review of a previous histopathological examination report showed the presence of ganglion cells and absent hypertrophic nerve fibers in the provided specimen (Figure 2).

A decision to close the stoma was made. In the postoperative period, the patient developed abdominal distension and did not pass stools but was passing flatus. We suspected impacted fecaloma in the rectosigmoid region; colonoscopy was performed, which revealed a diaphragm obstructing the lumen. The patient was again examined; the finding simulated a transition zone with a collapsed distal and dilated proximal segment (Figure 3). Colon was opened longitudinally at the transition zone and type I sigmoid atresia (windsock-like deformity) was found (Figure 4).

Excision of web and wing-shaped repair was performed and the previous anastomosis site was exteriorized. Postoperative recovery was uneventful. In the follow-up visit, a distal colostogram was performed and was normal; colostomy closure was subsequently performed.

3. Discussion

There are three anatomical types of CA; the least frequent is type I, which is characterized by a mucosal diaphragm

Figure 1  Barium enema film showing normal passage of contrast through rectum, sigmoid, and descending with slight narrowing at the level of sigmoid colon. No evidence of any mucosal irregularity, filling defect, or dilatation.

Figure 2  Photomicrograph of rectal biopsy specimen (40×) showing ganglion cells. No hypertrophic nerve fiber seen.

Figure 3  Operative photograph showing narrow rectum with dilated sigmoid and transition zone.
completely occupying the lumen without seromuscular interruption. These webs can have a small opening, thus preventing complete obstruction and often resulting in delayed diagnosis. HD is recognized in at least 2% of patients with CA. A study hypothesized that when a vascular insult occurs before the retroperitoneal fixation of the colon at 11 weeks of gestation, caudal migration of the myenteric nerve is interrupted. Therefore, it is imperative to rule out HD in every patient with CA.

Approximately 50–90% of children with HD present during the neonatal period with abdominal distension, bilious vomiting, and feeding intolerance. Plain abdominal radiographs generally show dilated bowel loops and the air fluid level throughout the abdomen. A differential diagnosis includes intestinal atresia, meconium ileus, meconium plug syndrome, and other less common conditions such as colonic atresia. A water-soluble contrast enema is generally administered as a first step in the diagnostic process. The salient findings on a contrast enema is the narrowing of a variable length of the distal bowel that has an irregular mucosal outline and involves the rectum and distal sigmoid colon with the reversal of the recto-sigmoid index and a funnel-like transition into a dilated proximal sigmoid and descending colon, and there is a retention of contrast in the colon on the 24-hour postevacuation film.

A diagnosis of CA is straightforward when facilitated by a water-soluble contrast enema, and preoperative contrast is recommended in neonates with distal intestinal obstruction. Early operative management is undertaken because of the risk of perforation and volvulus. Because of the association with HD, suction rectal biopsy for evaluating ganglionic cells should be performed in the operating room if primary anastomosis is planned. Similarly, if colostomy is appropriate, the myenteric plexus at the colostomy site should be examined during operation, and suction rectal biopsy should be performed before repair.

Type I atresia may exhibit a small perforation that prevents complete obstruction, leading to delay or misdiagnosis as in our case. A treatment decision may be inaccurate even after a barium enema, as in our case; however, a water-soluble contrast enema should be administered for diagnostic examination. In this patient, a preoperative contrast enema with a high index of suspicion for type I sigmoid atresia and a longitudinal colotomy at the point of caliber change during the first exploration would have prevented the morbidity associated with misdiagnosis. In addition, on the basis of suspicion on a contrast enema, colonoscopy before the closure of colostomy would have reduced the morbidity, even if diagnosis was missed at the first operation. Because of the rarity of this condition, a high index of suspicion is required to diagnose type I sigmoid atresia, particularly with perforation of the web.

References