Ro J Pediatr. 2023;72(4)
DOI: 10.37897/RJP.2023.4.4

Concurrence of Hirschsprung disease and annular pancreas: a rare case report

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ABSTRACT

Hirschsprung's disease (HD) and annular pancreas are both uncommon congenital gastrointestinal malformations. Although additional abnormalities are associated with 18% of HD cases, there is limited evidence of concomitant HD and annular pancreas. We present a full-term, two-day-old male with abdominal distended, and failure to pass first stool after 3 days of observation and rectal washout the abdominal X-rays showed a double bubble appearance. Intraoperative finding, the patient had pancreas annulare (PA). Colonic aganglionosis was discovered using full-thickness rectal biopsies (FTRB). At the age of seven months, a transanal endorectal pull-through was performed without complications. Although the diagnosis of these conditions might be difficult due to their clinical similarities, appropriate diagnosis, and surgical management are possible and can dramatically minimize morbidity and death in the newborn.

Keywords: Hirschsprung disease, annulare pancreas, gastrointestinal malformations, case report

INTRODUCTION

Congenital gastrointestinal abnormalities are infrequent and more prevalent in patients with genetic conditions [1,2]. Hirschsprung disease (HD) is a congenital neuroenteric disorder characterized by a failure of neural crest cell craniocaudal migration, resulting in various lengths of intestine aganglionosis [3-6]. HD has a diverse representation. Delay meconium within the first 24 hours of life may result in a missed diagnosis, with a subsequent diagnosis confirmed only after the emergence of constipation that persists [4,7,8]. HD affects around one in every five to ten thousand live births [3,4,9]. Most cases are solitary traits, while 18% are connected with additional congenital defects. The annular pancreas is defined by a pancreatic tissue band around the duodenum, which may cause obstruction and manifest in the same way as duodenal atresia. Its incidence has been estimated to be between 1 in 1000 and 3 in 20,000 [10,11]. Although all of these two disorders can be linked to Trisomy 21, HD that presents simultaneously with the annular pancreas is poorly understood [12]. We present a case of coexisting HD and annular pancreas without trisomy 21 [3,13].

CASE REPORT

On the second day of life, a 39-week-old male newborn was referred to the emergency department due to delayed meconium, distention of abdominal, and bilious vomiting. The patient was born to a 34-year-old G3P2003 mother by c-section due to breech presentation with the weight at birth being 3000 g. Physical examination revealed normal vital signs, bilious retention on an oral gastric tube, abdominal distension, visible venaectation, visible intestinal contour, and decreased bowel sounds (Figure 1). A digital rectal examination revealed normal sphincter tone, an ampulla that had not collapsed, and explosive meconium.

Large bowel dilatation was discovered on the first abdominopelvic radiography (Figure 2). But even after frequent rectal washouts, bilious vomiting and distension in the upper abdomen persist. There was a suspicion of obstruction at the level of

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Article History:
Received: 24 December 2023
Accepted: 28 December 2023

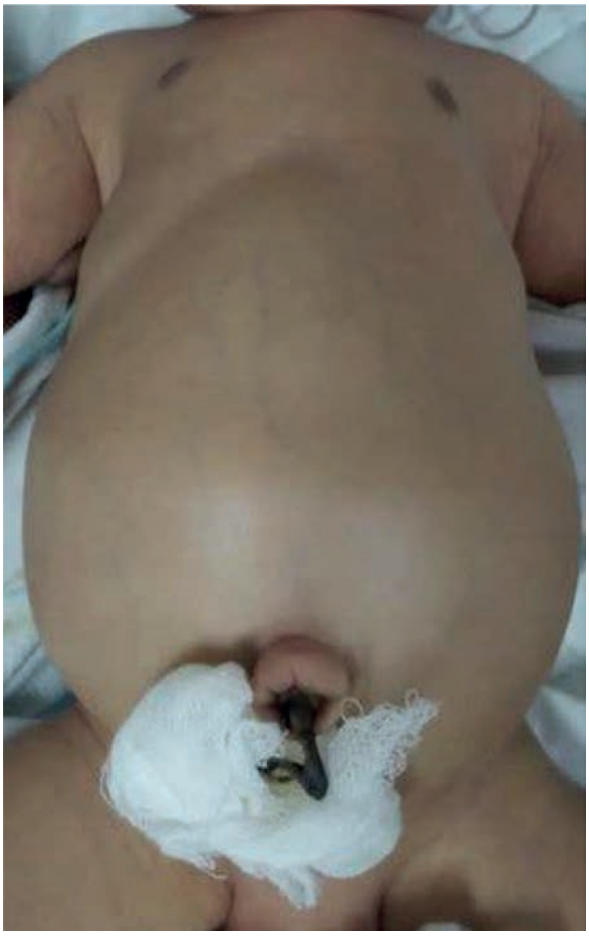

FIGURE 1. Clinical picture showing abdominal distension

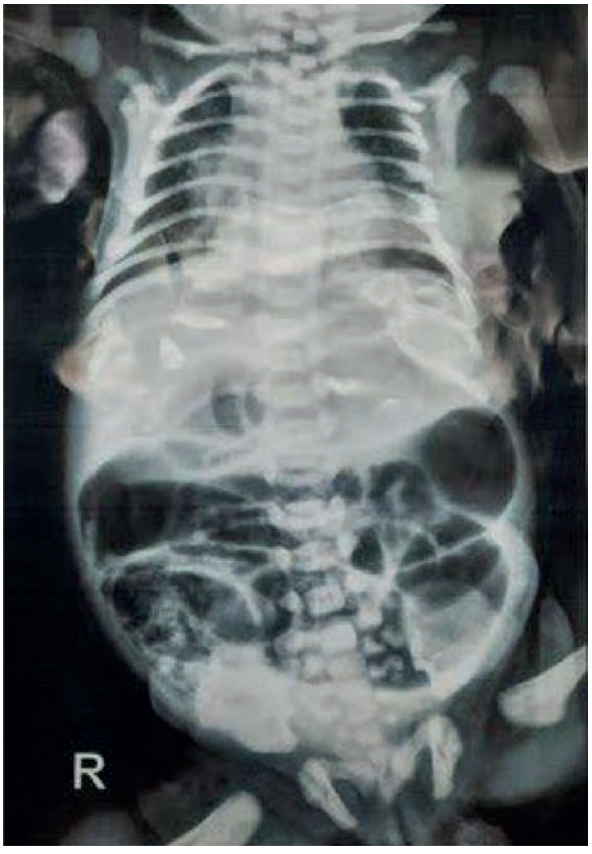

FIGURE 2. Initial abdominopelvic radiography revealed significant bowel dilatation

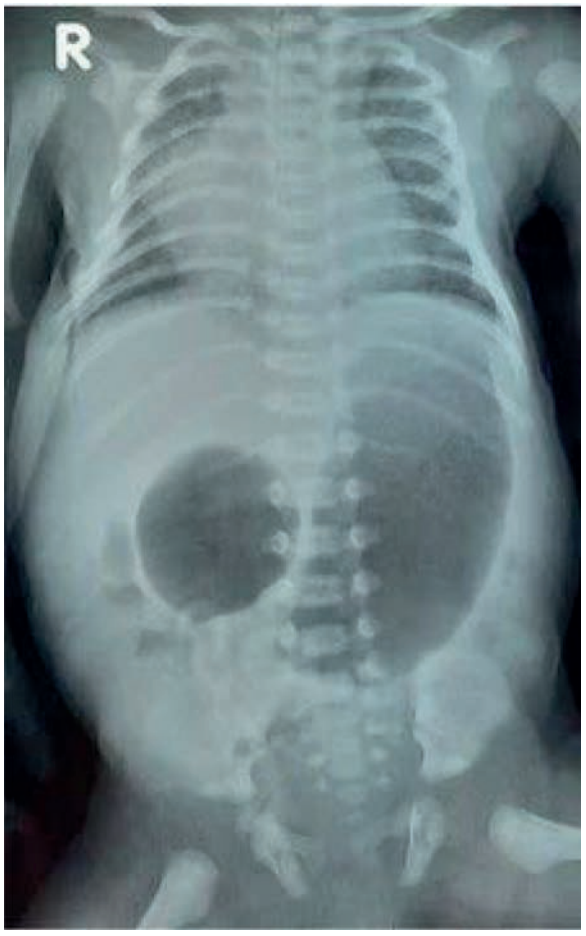

FIGURE 3. Abdominopelvic radiograph evaluation after washout revealed a double bubble in the mid-abdomen without distal bowel gas.

the duodenum. Abdominopelvic radiograph evaluation after washout revealed a mid-abdominal double bubble with minimal gas on the distal bowel (Figure 3). Due to diagnostic equipment limitations, an upper GI study could not be performed.

The patient's initial treatment was a rectal washout, but after complaints of persistent vomiting and distention in the upper abdomen and a radiological examination, it was decided to perform an exploratory laparotomy at three days old with suspicion of duodenal obstruction. Intraoperatively, a Kimura-type duodenoduodenostomy was used to repair an annular pancreas involving the second half of the duodenum (Figure 4). Full-thickness rectal biopsies (FTRB) were also performed which revealed colonic aganglionosis. The postoperative course was uncomplicated, with bowel function on the day tenth and a return to home after two weeks.

A transanal endorectal pull-through was performed at seven months of age with the uncomplicated postoperative course, and he was last seen in the clinic at one year old and weighed 10 kg with good nutritional status. To date, he has experienced no complications with HD or annular pancreas.

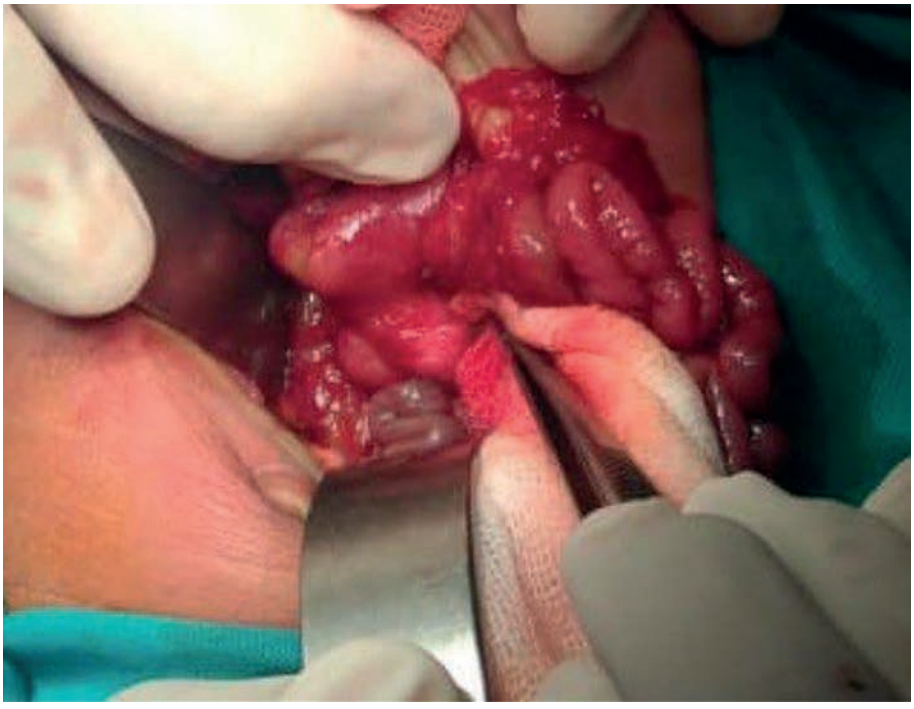

FIGURE 4. Annular pancreas at the laparotomy.

DISCUSSION

In the general population, HD and annular pancreas are uncommon. There have been multiple reports of HD and ileal atresia, some of which have been related to abnormalities such as gastroschisis and esophageal atresia [13-15]. Trisomy 21 is a further correlation between these abnormalities [16,17]. The current case had concurrent HD and annular pancreas but no trisomy 21. This was never reported before. It is important to highlight that the annular pancreas and other proximal obstructive gastrointestinal anomalies may be clinically hidden by HD distal obstruction.

It is difficult to explain the coexistence of various abnormalities because their pathophysiological mechanisms differ. Recent investigations using animal models to manipulate the Hedgehog genes have created results comparable to ours. Researchers produced mice models with Sonic hedgehog (Shh) or Indian hedgehog (Ihh) gene mutations in one study [18]. Hedgehog (Hh) signals are thought to play a crucial role in the development of the embryonic gut. Sonic Hedgehog (Shh) and Indian Hedgehog (Ihh) ligands are expressed in endoderm even before gut tube folding is complete, and ongoing expression of this pathway is required for several stages of gut organogenesis [19]. The molecular cause of frequent Gastrointestinal Tract Malformations (GTMs) has been linked to abnormal Hh signaling [20]. Shh and Ihh mutants both had intestinal malrotation and annular pancreas, Ihh mutants, on the other hand, had colonic aganglionosis, a phenotype comparable to HD, as well as other abnormalities [17]. These findings show that Hedgehog genes are critical in the patterning of the mammalian gut and that newborns with several concurrent gastrointestinal abnormalities could have a genetic abnormality.

It is crucial to highlight that distal obstruction from HD may disguise the annular pancreas and other proximal obstructive malformations of the gastrointestinal system. These discoveries should raise the surgeon's attention to the potential of HD and the annular pancreas. Recognizing the annular pancreas early allows for prompt surgical treatment, which improves long-term prognosis in infants with HD who also have an annular pancreas.

CONCLUSION

Although the coexistence of HD and annular pancreas is uncommon, the cases presented here show that it is a relevant differential diagnosis to investigate. As a result, any child who has had Hirschprung disease or vice versa should be evaluated for the annular pancreas. Prompt diagnosis and surgical intervention can reduce mortality and morbidity significantly.

Acknowledgments: the authors would like to acknowledge all the hospital staff who assisted us in treating the patient.

Conflict of interest: the authors declare no conflict of interest

Financial support: none declared

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