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Case Report

Paediatrics Section

Late Diagnosed Hirschsprung Disease: A Case Report

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ABSTRACT

Hirschsprung Disease (HD) is mainly a diagnosis of the neonatal period. It is mostly suspected within the first few days of life, the classical telltale sign is an infant not passing meconium. Paediatricians continue to encounter cases of late diagnosed HD. A late diagnosis of Hirschsprung poses an unnecessary burden and suffering on the child and family and increases the likelihood of long-term health complications for the patient. Hirschsprung-Associated Enterocolitis (HAEC) is one of the acute serious complications of late diagnosed HD. It is the leading cause of morbidity and is responsible for half of the deaths associated with Hirschsprung disease. Hence, HAEC should be kept in mind as a probable diagnosis by evaluating physicians particularly when making assessments for a child with possible necrotising enterocolitis or distal bowel obstruction with loose stools. This case report highlights the importance of keeping HD on the differential diagnosis of an ill-appearing child presenting with growth faltering below growth curve for age and in settings of chronic constipation. The patient discussed in this case evolved from an admission due to poor weight gain and fever to concerns for paediatric surgery evaluation. Her symptoms progressed to include not just clinical changes, but also physical examination reflected worsening abdominal exams. Patient underwent biopsy which showed aganglionic bowel segment which resulted in surgical resection. Patient had complete recovery without major complications at two weeks postsurgery.

Keywords: Abdominal distention, Constipation, Poor weight gain

CASE REPORT

A 14-month-old, early term (38 4/7 weeks) appropriate for gestational age female infant presented to the Emergency Department (ED) with complaints of fever and constipation. Mother reported intermittent fever for two days reaching a temperature of 105° F at home, measured rectally with poor response to acetaminophen. As per patient's grandmother, the patient had not passed stools in more than a week and the stomach felt “rock hard” associated with decreased oral intake for two days.

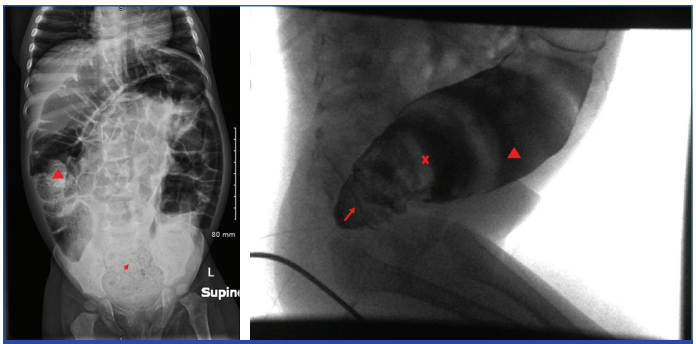
Prior to the fever, the patient had 3-4 watery, liquid, yellow stools without any blood for three days. The mother reported decreased activity level the day before the Emergency Department visit and decreased dietary intake. Family decided to bring the patient to hospital after three episodes of non bloody, non bilious emesis. The number of wet diapers remained the same as her normal pattern.

Absence of cough, nasal congestion, vomiting, rash, new foods was reported. Any sick contacts or recent travel were denied. Mother of the infant stated that the patient had previously been taking 7.5 mg of polyethylene glycol daily since the age of 6 months due to chronic constipation but had recently stopped. Upon arrival to the hospital, patient's vitals were- weight 7 kg (0.43% percentile,: World Health Organisation Child Growth Standards for Girls 0-2 years, growth chart) [1], temperature 103.3°F (measured rectally), heart rate 148 beats/min, respiratory rate 38 breaths/min.

She was tired looking and crying, without signs of distress. On examination, she had dry mucous membranes, abdomen extremely distended, firm and tender to palpation. Bowel sounds were present and no organomegaly was appreciated, bowel loops were palpable. The patient was found to have an empty rectal vault on a rectal exam with no forceful stool expulsion. Abdominal x-ray showed significant stool in the distal colon and rectum with prominent gaseous distention of the remaining colon [Table/Fig-1].

Patient received a tap water enema and was admitted to the inpatient ward due to concerns regarding poor weight gain, fever to rule out serious bacterial infection and abdominal distension. At this point, there was a broad spectrum of differential diagnoses such as rule out intussusception, small bowel obstruction, toxic megacolon, fecal impaction with superimposed viral gastroenteritis, including possibility of enterocolitis in suspicion of Hirschsprung disease.

Over the course of her first day of admission, fever was controlled with alternating oral acetaminophen 15 mg/kg and ibuprofen 10 mg/kg every six hours as needed. Paediatric gastroenterology was consulted and recommended to continue with oral polyethylene glycol 7.5 mg daily and to add oral lactulose 15 mL twice per day. The patient did not tolerate polyethylene glycol or lactulose orally due to an episode of non bloody, non bilious vomiting and placed on Nil Per Os (NPO). Tap water enema was done twice, producing some pasty stools. Serial abdominal exams were done pertinent for absent bowel sounds and tenderness to palpation. There was an increase in her abdominal girth from 49 cm to 51.5 cm in less than 24 hours since admission. The findings on abdominal exam and increasing abdominal distention prompted paediatric surgery consultation. It was therefore recommended that the patient should be started on intravenous piperacillin-tazobactam 100 mg/kg every 8 hours and intravenous metronidazole 10 mg/kg every 8 hours.



[Table/Fig-1]: Moderate stools in colon (red arrow head) and rectum (red arrow). Gaseous distention along the large intestine. **[Table/Fig-2]:** Contrast barium enema. Reversal of rectosigmoid index and mucosal irregularity. Transition zone (red x) demonstrated by small caliber rectum (red arrow) in comparison to sigmoid (red arrow head). (Images from left to right).

Rectal irrigation with normal saline via rectum through 16-French Foley catheter was started promptly. The patient underwent a contrast enema study, which showed a reverse rectosigmoid ratio, suggesting Hirschsprung disease [Table/Fig-2]. Patient continued NPO and sustained multiple rectal irrigations, which significantly improved her abdominal distention.

She underwent a total thickness rectal biopsy in which no ganglion cells were identified confirming the diagnosis of Hirschsprung disease. The pathology report for the rectosigmoid resection showed complete absence of distal ganglion cells. Shortly after histology and pathology results were obtained, she underwent trans-anal rectal pull-through (Soave's procedure) which was first designed to treat HD by Dr. Soave, Italian surgeon [2]. Patient had complete resection of about 15 cm long segment of bowel above dentate line on rectum. This length was based on frozen section sent for pathology from the rectosigmoid junction and colon while in the operating room. The colon was then approximated and anastomosed to dentate line on rectum. Digital rectal examination was done to assess for good anal tone and confirm patent anastomosis.

The patient was continued on intravenous ceftriaxone 50 mg/kg every 24 hours and metronidazole 7.5 mg/kg every 8 hours after surgery. Careful instructions were given for no anal examination, no touching or wiping near anal area. On postoperative day 2, antibiotics regimen was revised, ceftriaxone was discontinued, and the patient was advanced to regular diet after having multiple bowel movements. She was discharged on postoperative day 4 in good condition, tolerating regular diet, regular bowel movements and pain well-controlled. Patient was sent home to complete 14 days of oral metronidazole 13 mg/kg every 8 hours and oral acetaminophen 15 mg/kg as needed.

On two weeks follow-up after the surgery she developed candida diaper rash. Patient was prescribed nystatin 100000 unit/gm ointment twice daily to diaper area for 10 days. She reported multiple soft and brown bowel movements. Her appetite was back to baseline.

DISCUSSION

Hirschsprung disease is mainly a diagnosis of the neonatal period. It is mostly suspected within the first few days of life, classically by the telltale signs of an infant not passing meconium in the first two days of life. In 80% of cases, HD is diagnosed within the first year of life, with 50-60% of patients diagnosed within the first month of life [3]. This disorder is uncommon in adolescence and adulthood. When present in this age group, it shows up as a short or an Ultrashort-Segment Disease (USHD) of aganglionic segment of bowel [4].

Though accounting for less than 10% of the cases, paediatricians encounter issues of late-diagnosed HD. The first reason for the delayed presentation of HD is inadequate access to specialized centers and delayed referrals. The second reason is that only 90% of aganglionosis produces clinical signs and symptoms of HD [5]. Thus, leading to missed diagnosis and later presentation as faltering growth due to protein-losing enteropathy. Mild HD cases can be underdiagnosed, either due to lack of symptoms or because symptoms are mistaken with other conditions. A certain degree of constipation of fluctuating intensity is not uncommon. In such cases, fecaloma characterising clinical progression, often associated with nutrition and growth changes, is commonly misinterpreted as functional constipation [6]. As a clinician, few differences could help differentiate the latter from HD. According to the book, Nelson's textbook of paediatrics [7], HD disease onset is at birth whereas functional constipation usual onset is after 2 years of age. Another differing point between the two is that in HD there is no sphincter relaxation which was not part of the investigations done for this case. Another important difference is that when barium enema is done, functional constipation has no transition zone but in HD a zone where there is a visible change in the diameter or caliber of the loop of bowel between rectum and colon can be seen as also noticed in the present case. The level of colonic caliber change in a contrast enema can help to predict the degree of aganglionosis and therefore the length of the colonic segment to be resected and whether laparoscopic assistance during surgery is necessary or an open colectomy is indicated [8].

Part of the misdiagnosis could also be attributed to HD's varied presentation, classified according to the extent of the aganglionic area as a short segment, long-segment, total colonic, and USHD. When comparing HD and USHD, the latter has ganglion cells on rectal biopsy and no transition zone on barium enema. Furthermore, neither USHD or classical Hirschsprung's disease have reflex internal sphincter relaxation on rectal manometry [9].

Hirschsprung-Associated Enterocolitis (HAEC) was first identified in the later part of the nineteenth century by Härald Hirschsprung [10]. HAEC constitutes intestinal inflammation defined by the following clinical features: fever, abdominal distention, diarrhea, and sepsis which were all present in the case in question [11]. Hirschsprung highlighted important histological findings: crypt abscesses, mucosal ulceration, and transmural necrosis which aid in the diagnosis of HAEC [12]. HAEC is the most feared complications associated with Hirschsprung disease as it is responsible for half of deaths in patients with this condition [13].

Some patients reach adulthood without a diagnosis for this disease. Typically, patients go to the doctor with a history of constipation requiring frequent laxative or enema use [14]. The current frequency of the disease in adults is unknown, especially since HD is an overlooked and misdiagnosed illness in this age group. Other factors contributing to a delayed HD presentation include illiteracy, parental ignorance, and poverty [15]. The hallmark of late-diagnosed HD is chronic constipation without encopresis as far as the newborn period [16]. On physical examination, patients with late-diagnosed HD often have abdominal distention and palpation of large fecaloma.

Management of HD, early or late diagnosed is mainly surgical. In this case, the patient underwent transanal Soave pull-through which has both advantages and disadvantages. The positive factors in this case will be short operation time, minimal bleeding and decreased morbidity in comparison to transabdominal pull-through surgery [17]. Even postsurgical repair, patient can still develop HAEC with an incidence of up to 40% [18].

Also, in the case of late-diagnosed HD malnutrition and anemia are common therefore nutrition needs to be addressed. Patients with late-diagnosed HD are predisposed to severe complications such as intraoperative bleeding, frequent bowel movement, temporary fecal incontinence with the negative effect on mental health, inadequate anastomosis healing, soiling and lastly mortality due to enterocolitis [15]. Constipation and incontinence are the most important markers to assess the outcome of the patients. In the present case, at two weeks follow-up postoperative procedure pull-through Soave did not have constipation and had a normal number of bowel movements per day which were described as soft without any discomfort.

CONCLUSION(S)

Although commonly diagnosed in patients younger than 12 months, Hirschsprung disease should be a differential diagnosis when a child presents with poor weight gain, fever and chronic constipation. The late diagnosis of Hirschsprung poses unnecessary burden and suffering on the child and family and increases the likelihood of long-term health complications on the patient. The case presented showed distended abdomen, radiographic images with concerns for bowel obstruction increased the suspicion for HD. The contrast enema showing the transition zone is the pathognomonic clue to achieve the diagnosis of late onset Hirschsprung disease.

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