

An Unexpected Case of Chronic Constipation in A 16-Year-Old Girl Turned Out to Be Hirschsprung's Disease

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ABSTRACT

Background: Constipation is a disorder in the gastrointestinal tract, which can result in the infrequent stools, difficult stool passage with pain and stiffness. A large sum of public funds is also being spent on caring for children with constipation due to repeated hospital admissions, emergency room visits, and regular clinic visits because of recurrent exacerbations of their symptom. **Case:** An older children 16-year-old girl complaining of not having smooth bowel movements since birth. She complaints of nausea, vomiting, and flatulence are present. The stomach is minimal enlarged, but stomach pain is absent. the patient underwent a barium enema colon examination and underwent a punch biopsy operation which confirming the diagnosis of Hirschsprung's disease. The patient underwent trans endorectal pull-through (TERPT). Several weeks later, the patient returned to the pediatric surgery clinic for a follow-up, and it was found that the patient's intestinal function was normal. There were no complaints of vomiting, bloating, or abdominal pain. **Conclusion:** Chronic constipation in older children cannot rule out the possibility of a diagnosis of Hirschsprung disease. Medical history alarm symptoms or signs in children differ according to the age of onset of symptoms. A series of tests should be performed to exclude underlying diseases. so that the chosen treatment can become definitive therapy.

Keywords: constipation; older children; Hirschsprung's disease

INTRODUCTION

Constipation is a disorder in the gastrointestinal tract, which can result in infrequent stools and difficult stool passage with pain and stiffness. Acute constipation may cause closure of the intestine, which may even require surgery. history and physical examination can be considered the main initial approaches. Many definitions are described by using self-reported constipation and formal criteria. [1]. Chronic constipation is a common and persistent problem in childhood, accounting for approximately 3– 5% of visits to pediatric outpatient clinics and 10– 20% of visits to pediatric gastroenterology clinics. Although 40–50% of childhood cases respond to drug treatment, in many children constipation remains refractory to drug therapy. In these cases, all organic causes for constipation such as anatomical (colonic stenosis, duplication, etc.) or neurological (Hirschsprung's disease (HD), intestinal neuronal dysplasia (IND), etc.) abnormalities, endocrine and metabolic causes should be considered [2].

Children with constipation are referred for a rectal biopsy when a suspicion of Hirschsprung's disease (HD) has been raised. To diagnose HD, rectal biopsies are examined for the presence of ganglion cells. The biopsies are classified as positive for HD (a ganglionic) if no ganglion cells are detected after examination of a sufficient number of histological Sections. Often supplemented with immunohistochemical staining and/or enzyme histochemistry [3].

Trans anal pull-through for Hirschsprung's disease (HD) is becoming the preferred treatment option worldwide. This technique has been practiced in Nigeria for over 15 years but less so in sub-Saharan Africa.[1] An important benefit of trans anal pull-through is the absence or minimal abdominal manipulations, especially when combined with laparoscopy or laparotomy. Laparoscopy as an effective tool for colonic biopsy, identification of transitional zone, and mobilization of the colon is well established[4].

CASE REPORT

A 16-year-old girl was brought by her parents to the pediatric surgery clinic with a referral from a Level One Health facility, complaining of not having smooth bowel movements since birth. The patient has a habit of defecating once a week, sometimes even once every ten days. This complaint is felt to be more severe if the patient does not eat enough fiber. Complaints of nausea, vomiting, and flatulence are present. The stomach is enlarged, but stomach pain is absent. Initially, the patient was given laxatives after every visit to the general practitioner to facilitate defecation. However, for several months, despite taking laxatives, the patient still finds it difficult to defecate. The patient's previous medical history of late meconium passage was denied, and there was no abdominal enlargement at birth. Menstrual disorders were also denied. Physical examination did not reveal any signs of acute abdomen, including abdominal pain or distension. Vital signs are within normal limits. Rectal examination did not reveal any abnormalities.

Finally, the patient underwent a barium enema colon examination to assess the distal intestinal passage. The results showed that the entire colon was dilated, the mucosa was smooth, the haustra were still visible, and there were no filling defects.

The diagnosis indicated colonic dilatation, suggestive of megacolon due to peristaltic disorders, specifically Hirschsprung's disease. Three days later, the patient underwent a punch biopsy operation in the recto rectal area due to suspected abnormalities in muscle and nerve function. The anatomical pathology examination results revealed that the submucosa and tunica muscularis showed plexuses, and no ganglia were observed, confirming the diagnosis of Hirschsprung's disease.

Several weeks later, the patient underwent scheduled trans endorectal pull-through Soave surgery. During the operation, a transition zone was discovered in the rectosigmoid section, prompting the patient to undergo Trans Endorectal pull-through (ERPT). Tissue samples from the pulled-through area were examined in the anatomical pathology laboratory, revealing that the tissue had reached the nerve limit of the intestine. The patient remained hospitalized for three days and was discharged in stable condition.

Several weeks later, the patient returned to the pediatric surgery clinic for evaluation. It was determined that the patient's bowel function had normalized, with no complaints of vomiting, bloating, or abdominal pain.



FIGURE 1: Abdominal Distension of The Patient.

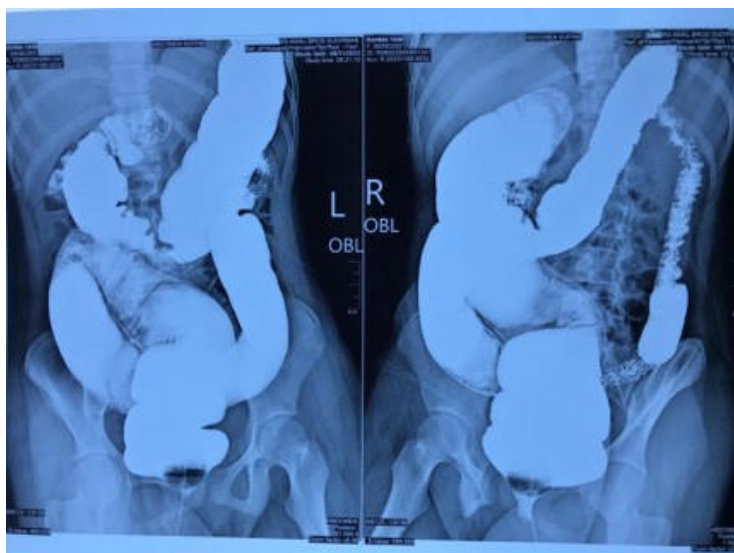


FIGURE 2: Barium Enema Depicts the Megacolon Due to Peristaltic Disorders.

DISCUSSION

Difficulty in defecation, with or without soiling, is often encountered in children. It presents a management problem for general practitioners, and parental concern is often high [5]. Childhood functional constipation (FC) is characterized by the presence of infrequent, and painful bowel motions, fecal incontinence, stool-withholding behavior, and occasional passage of large-diameter stools. A large sum of public funds is also being spent on caring for children with constipation due to repeated hospital admissions, emergency room visits, and regular clinic visits because of recurrent exacerbations of their symptoms [6].

Medical history alarm symptoms or signs in children differ according to the age of onset of symptoms which include a history of delayed meconium passage, onset before one month old, family history of Hirschsprung disease, hypothyroidism or coeliac disease, bloody stool, fever, bilious vomiting, or ribbon stool [7]. Alarm signs also include physical examination (including digital rectal examination) that detects failure to thrive, severe abdominal distention, tight and empty rectum, abnormal cremasteric or anal reflex, abnormal gluteal cleft or position of the anus, anal scars or fissures, major fear of per rectal exam, or anal hematoma [7]. A series of tests should be performed to exclude underlying diseases. Barium enema, endoscopy, anorectal manometry, and balloon expulsion tests can be considered to diagnose chronic constipation [8].

Hirschsprung disease (HD) or congenital aganglionic megacolon is an intestinal motor disorder, which is caused by the failure of neuroblasts originating from the neural crest (precursors of enteric ganglion cells) to migrate completely during intestinal development in the first 12 weeks of gestation. It shows an absence of ganglion cells in the submucosal and myenteric neural plexuses in a variable bowel segment causing a functional obstruction and mainly producing the symptoms of constipation. The other symptoms that lead to an early suspicion of HD are defined in the newborn as a greater than 48-hour delay in the elimination of meconium associated with abdominal distension and vomiting [9].

HD is mostly confined to the rectum (79.8%), followed by the recto-sigmoid junction (12.5%) and the descending colon (0.8%). A markedly dilated proximal colonic segment with a transition zone and a narrowed distal colonic segment on CT and double-contrast barium enema suggest adult HD. If available anorectal manometry should be performed. The latter is however the gold standard; accuracy depends on the biopsy site, the representativeness of the tissue, the number of specimens, and the pathologist's skill. The specimen can be obtained via suction, endoscopy, or open full-thickness surgical rectal wall excision in the operating room [10]. The genetics of HSCR are complex and involve multiple susceptibility loci. The summary genetic view of HSCR susceptibility is that it arises primarily from the segregation of alterations in multiple genes, both rare and common, most of which impair signal transduction by the RET receptor tyrosine kinase [11].

Clinical management of constipation has several facets. The main approaches are non-pharmacological interventions (education and demystification, dietary adjustment, toilet training, behavioral interventions, use of biofeedback, and pelvic floor physiotherapy), pharmacological interventions (oral and/or rectal laxatives, including novel drugs such as prucalopride and lubiprostone), and surgical interventions (antegrade enema and bowel resection), and other novel modalities, such as neuromodulation. [12]. The surgical management of HD involves employing various techniques. These techniques are widely practiced worldwide and commonly include procedures such as Swenson, Duhamel, and Yancey-Soave. Currently, the two most frequently performed surgical procedures to treat rectosigmoid HD are approaches. Surgery for HD aims to restore normal bowel function. Endorectal pull-through (ERPT), which includes both total trans anal (TERPT) and laparoscopic endorectal pull-through (LERPT) TERPT is a preferred technique for HD, replacing staged procedures. TERPT and LERPT are commonly performed for rectosigmoid HD [13]. Furthermore, primary or staged surgery for HD remains controversial. Presently, most doctors adopt the one-staged operation for HD patients due to the safety and efficacy of the procedure [14]. Older children may also undergo a preoperative period of rectal irrigations in order to attempt to decompress the bowel. If this is not successful, often an ileostomy is performed in order to divert the fecal stream and allow for decompression, prior to attempting a pull-through procedure. [15].

The structured follow-up to adulthood, including the transition of care, is indicated in HSCR. As bowel dysfunction is most common during the first few years after surgery, patients should be monitored more closely for the early detection of problems, including defective continence, enterocolitis, and late complications [16]. Rintala score or Krickbeck classification can be used to determine the functional result. The score is based on seven parameters that evaluate social issues, defecation frequency, soiling the incidence, ability to describe the urge to defecate, and ability to retain feces. Evaluation of bowel movements, constipation, and soiling is conducted using the Krickbeck criteria [17].

CONCLUSION

In cases of chronic constipation in older children, the possibility of a diagnosis of Hirschsprung's disease cannot be ruled out. Medical history of alarm symptoms or signs in children differing according to the age of onset of symptoms, including a history of delayed meconium passage, should also be obtained. A series of tests should be performed to exclude underlying diseases. Tests such as barium enema, biopsy, anorectal manometry, and balloon expulsion test can be considered. This comprehensive examination is crucial so that the chosen treatment can become definitive therapy.

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