Total collectomy in adult woman with Hirschsprung’s Disease: a case report

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Abstract

Hirschsprung’s disease (HD) is a congenital anomaly of innervation of the lower intestine, limited to the colon, resulting from partial or total obstruction. The main symptom is the inability of a newborn to evacuate within 48 hours after birth, which can cause intestinal obstruction in the neonate and megacolon in children and adults. Therefore, this study reported the case of a 30-year-old female patient with HD affecting the entire colon, where we opted for surgical treatment. In the present study, it was emphasized the medical need to have a better understanding of the clinical picture of HD, as well as its complications and the importance of early diagnosis. In the report presented, it was discussed a late diagnosis of HD and how it affects the patient’s life. It presented the improvement in the quality of life that can be obtained by offering an accurate diagnosis and adequate treatment for the patient.

Keywords: Hirschsprung’s disease. Congenital megacolon. Obstruction.

Introduction

Hirschsprung’s Disease (HD), also known as Congenital Megacolon, was described by Harold Hirschsprung in 1888 as a congenital disorder in a newborn with severe constipation associated with colonic dilatation and hypertrophy [1-3]. This disease has an embryonic origin between the 5th and 12th week of gestation. During this period, the undifferentiated cells of the vagal and lumbosacral neural crest migrate to the gastrointestinal tract to form the submucosal myenteric plexuses, with the need to multiply, survive and complete their differentiation [1,4].

When there is any change in this system, neural crest cells do not reach the colon or do not complete their differentiation, generating an aganglionic intestinal segment [1]. Approximately 80 to 90% of cases are classified as short-segment HD (affecting the rectosigmoid region distally), 25% are ultra-short-segment (affecting the distal anorectal region), and the long segment is rare and may extend into the small intestine [2,5].

The incidence of this disease is about 1:5,000 live births, and it affects males in a proportion of 4:1 [5,6]. The newborn usually has an acute obstructive abdomen or constipation, associated with abdominal distention and paradoxical diarrhea. In older children, the most common form of presentation is chronic constipation [2,7,8].

To elucidate the diagnosis, in addition to the clinical picture, it is necessary to carry out complementary exams, among them: simple radiography of the abdomen (dilated bowel loops that suggest obstruction); barium enema to define the transition zone of the aganglionic segment, with decreased caliber of the rectum, sigmoid and enlarged colon proximal to this portion; anorectal manometry (absence of the rectosphincteric reflex). Although there are no pathognomonic radiological signs, the definitive diagnosis is obtained by histopathological study, the gold standard being rectal biopsy [8-10].
The treatment of this disease is always surgical, aiming at removing the aganglionic part and restoring the continuity of the intestine \[2,10\]. Therefore, this study reported the case of a 30-year-old female patient with HD affecting the entire colon, where we opted for surgical treatment.

**Methods**

**Study Design**

The present study was elaborated according to the rules of the CARE case report (https://www.care-statement.org/). A descriptive literature review was also carried out to provide sufficient scientific data for the theoretical basis of this study. The patient's medical record was analyzed, and who authorized access to it and signed the Free and Informed Consent Term, together with those involved in this work. This patient underwent treatment and medical follow-up with the supervisor of this work. A thorough evaluation of the aspects inherent to the physical examination, complementary exams, and surgical technique was carried out, to correlate with the literature cited in the bibliography. Data collection and analysis of the patient's medical records were performed at Hospital Unimed São Domingos Catanduva, São Paulo.

**Ethical Approval**

This study was analyzed and approved with the number 5.701.687 by the Research Ethics Committee from the FAMECA/UNIFIPA, Catanduva, Brazil, and obtaining the Informed Consent Form according to CNS/CONEP Resolution 466/12.

**Case report**

**Patient Information and Clinical Findings, Timeline, Diagnostic Assessment, Therapeutic Intervention, and Follow-up**

Patient A.F.D.M, female, 30 years old, only hypothyroidism as comorbidities in control with levothyroxine sodium 100 mcg daily. She sought our care with a complaint of abdominal pain accompanied by a small amount of bowel movement since childhood, alternating with periods of constipation, staying up to 12 days without bowel movement associated with the recurrent use of suppositories and/or enemas.

She mentioned that in the last 13 years she underwent multiple consultations with CT scans and colonoscopies, however without assistance in the diagnostic elucidation. In 2020, she underwent a consultation where an anorectal manometry exam was requested, with the result concluding the presence of reduced voluntary contraction and paradoxical contraction of the puborectal muscle. Therefore, on this occasion, she was diagnosed with megacolon and was instructed to seek a surgeon with experience in the surgical treatment of the pathology in question.

She also reported that he had two episodes of volvulus with spontaneous resolution in the last two years, the first in August/2021 and the last in January/2022. Shortly after the second episode of volvulus, the patient underwent treatment with our team, at which time a barium enema exam was requested, which showed signs of dilation and partial loss of haustrations from the splenic angle from the colon to the rectum (Figure 1).

![Image of imaging exam with a diagnosis of megacolon.](image-url)
Because of the diagnosis of megacolon due to Hirschprung's disease, it was opted for surgical treatment, in which total colectomy with ileorectal anastomosis was performed (Figure 2).

The patient did well in the postoperative period, with mild pain that was controlled with simple analgesics, good diet acceptance, and evacuation on the third postoperative day. In the third postoperative period, she presented well, with the criteria for hospital discharge.

She reported that since the surgery she has been pain-free, with good food acceptance, initially having liquid stools about eight times a day and progressively progressing to soft stools about three times a day. Denies complaints. She mentioned that her 4-year-old daughter also has difficulty in evacuating and has started follow-up for etiological investigation. However, she tested negative for megacolon.

The anatomopathological result showed a sometimes thin colonic wall where ganglion cells were not identified in the submucosal plexus in all the removed segments. In the myenteric plexus, ganglion cells are present in some cells. Immunohistochemical examination was positive for calretinin and PHOX2B in ganglion cells.

Figure 2. Total colectomy with ileorectal anastomosis.

Discussion

HD is a congenital anomaly that affects 1 in 5,000 live births and is predominantly 4:1 in males [11-13]. It is characterized by the absence of ganglion cells, and 80 to 90% of cases are diagnosed in the neonatal period [14]. Furthermore, in a recent study, it was highlighted that this disease has a familial predisposition, identifying a mutation of the alpha 9 protocadherin gene that is predominantly expressed in the myenteric plexus in human colon tissues [13]. In approximately 80% of cases, signs and symptoms began within the first month of life, and half of the children had the diagnosis confirmed after six months of age [2].

The delay between the onset of clinical manifestations and the investigation of the disease increases the risk of complications, with enterocolitis being the most frequent, in addition to increasing the morbidity and mortality of congenital megacolon [2]. The pathogenesis of this complication remains poorly understood. However, one of the theories states that the obstruction leads to bacterial stasis leading to their proliferation. When added to intestinal dilation and stretching of its walls, it impairs blood flow to the mucosa and the consequent increase in permeability with bacterial translocation [1].

The most frequent manifestations of enterocolitis consist of abdominal dilatation, constipation, vomiting, and delayed passage of meconium [10,11]. In one study, neonates had 90% abdominal distention, 67.1% vomiting, 23% constipation, and 51% of cases with delayed meconium elimination (> 48 h of life). In children after the neonatal period, constipation occurred in 68.7%, abdominal distension in 64.2%, vomiting in
37%, and delay in the elimination of meconium in 40.6% [2].

Most researchers have concluded that in children in the neonatal period there is a higher prevalence of obstruction, occurring in 42 to 81% of cases, while in children after one month of life constipation is predominant, occurring in 13 to 69% of cases [2]. A plain abdominal x-ray may show an enlarged small intestine or proximal colon. Available diagnostic tests include contrast enema, anorectal manometry, full-thickness rectal biopsy, and rectal suction biopsy [9,14].

The treatment is done through a surgical procedure in which the objective is to remove the aganglionic segment and reconstruction the intestinal transit [1]. When total organ involvement occurs with or without the involvement of the initial part of the small intestine (ileum), there is total colonic aganglionosis (TCA). It is characterized by being a rare form of HD. HD is fundamentally a disease of the neonatal period or early childhood. Late diagnosis leads to a worsening in the evolution of the condition as well as a decrease in the quality of life [15,16].

It is noted that even when the entire affected segment can be removed, there is no complete recovery of the patients, thus, it is necessary to better understand the pathophysiology of the disease to improve the therapeutic techniques, aiming at the total cure of the patient, reducing the risk of sequelae [2].

Conclusion

In the present study, it was emphasized the medical need to have a better understanding of the clinical picture of HD, as well as its complications and the importance of early diagnosis. In the report presented, it was discussed a late diagnosis of HD and how it affects the patient's life. It was presented the improvement in the quality of life that can be obtained by offering an accurate diagnosis and adequate treatment for the patient.

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Ethics approval

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Informed consent

Was applied.

Data sharing statement

No additional data are available.

Conflict of interest

The authors declare no conflict of interest.

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