**Hirschsprung's disease: case report**

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**Abstract**

**Introduction:** Hirschsprung’s Disease (DH), also known as Congenital Megacolon, has been described as a congenital disorder in Newborns (NB) with severe constipation associated with dilatation and hypertrophy of the colon. To elucidate the diagnosis, it is necessary to perform complementary tests, but the gold standard is rectal biopsy. The treatment is always surgical, aiming at the removal of the aganglionic part and the restoration of the continuity of the intestine. Thus, we chose to report a case of a 1-year-old and 10-month-old patient with chronic constipation from birth, who was diagnosed with DH, requiring surgical intervention.

**Case report:** A 29 days of life male patient comes with a complaint that he has been evacuating in small quantities since birth, with feces that look yellowish, pasty and with a characteristic odor, intercalating with periods of constipation. Mother reports that the RN remained for up to a week without evacuating from birth. With the DH hypothesis, an x-ray of the abdomen, an opaque enema, an abdominal distension and paradoxical diarrhoea, the newborn was hospitalized and underwent abdominal distension and paradoxical diarrhoea to investigate the disease. The delay between the onset of clinical manifestations and the investigation of the disease increased the risk of complications, with more frequent enterocolitis, in addition to increasing the morbimortality of the congenital megacolon.

**Discussion:** HD is a congenital anomaly that affects 1 in 5,000 live births. It is characterized by the absence of ganglion cells, and 80 to 90% of the cases are diagnosed in the neonatal period. The delay between the onset of clinical manifestations and the investigation of the disease increases the risk of complications, with more frequent enterocolitis, in addition to increasing the morbimortality of the congenital megacolon.

**Conclusion:** With the present study, we aimed to emphasize the need to alert the pediatrician to a better understanding of the clinical picture of HD, as well as its complications and the importance of performing the diagnosis early.

**Keywords:** Enterocolitis, Hirschsprung’s disease, Rectal biopsy

**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 colon dilatation and hypertrophy. [1-3] This disease has an embryonic origin between the 5th and 12th week of pregnancy. 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, the 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 intestine slender [2.5].

The incidence of this disease is about 1: 5000 live births, and affects males in the ratio of 4: 1. [5,6] The newborn usually presents with an acute obstructive abdomen or intestinal constipation, associated with abdominal distension and paradoxical diarrhea. In older children, the most common form of presentation is chronic constipation [2.8].
To elucidate the diagnosis, in addition to the clinical picture, it is necessary to carry out complementary exams, among them:

- Simple abdominal radiography (dilated intestinal loops that suggest obstruction);
- Opaque enema to define the transition zone of the aganglionic segment, with a decrease in the caliber of the rectum, sigmoid and an enlarged colon proximal to that portion;
- Anorectal manometry (absence of the rectosphincter reflex). Although there are no pathognomonic radiological signs, the definitive diagnosis is obtained by histopathological study, with 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]. With this, it was decided to report a case of a 1-year-old and 10-month-old patient with chronic constipation since birth, who was diagnosed with HD, requiring surgical intervention.

**Case report**

Male patient, 29 days old, enters the emergency room with a complaint that he was evacuating in small quantities since birth, with yellowish, pasty and characteristic odor stools, alternating with periods of constipation. Mother reports that the newborn remained for up to a week without evacuating since birth. Since then, Simethicone has been prescribed where small amounts of feces were released, with only “blotting” of the diaper. On physical examination, he had a globose abdomen with hydro-air noises present, hyper-panic and diffuse superficial palpation with pain. Rectal stimulation was performed with a number 16 probe, generating explosive and explosive feces output, with symptomatic prescriptions and emergency discharge.

After one year, the patient returned with chronic constipation, having undergone several pediatric consultations and unsuccessful treatment for constipation. In the simple radiography of the abdomen, dilated intestinal loops were observed that suggested intestinal obstruction (figure 1).

A biopsy was indicated with a histological study showing the absence of ganglion cells in the distal, proximal and sigmoid rectum. With the results of laboratory and imaging tests, the patient was diagnosed with HD and suggested surgical treatment. Initially, loop colostomy was performed and then, a rectosigmoidectomy with colon lowering.

For seven months, the patient evolved well without significant changes, with a good-looking anorectal anastomosis and a working loop colostomy. Due to the good evolution, the gastrointestinal transit reconstruction was performed with enteroanastomosis. There were no complications in the postoperative period, the patient evolved with evacuation present and then, outpatient follow-up.

**Discussion**

HD is a congenital anomaly that affects 1 in 5,000 live births and is predominantly 4: 1 in males. [12,13] It is characterized by the absence of ganglion cells, and 80 to 90% of cases are diagnosed in the neonatal period. [14,15] In addition, in a recent study it was highlighted that this disease has a familial predisposition, identifying a mutation of the alpha 9 protocaderin gene that is predominantly expressed in the myenteric plexus in the tissues of the human colon. [16] In approximately 80% of cases, signs and symptoms started in the first month of life, and half of the children had their 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, the most frequent being enterocolitis, in addition to increasing the morbidity and mortality of the congenital megacolon. [2,18] The pathogenesis of this complication remains poorly understood. [17,19] However, one of the theories states that the obstruction leads to bacterial stasis leading to their proliferation. When added to the intestinal dilation and the elongation of its walls, it impairs the blood flow to the mucosa and the subsequent increase in permeability with bacterial translocation [19].

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**Figure 1:** Abdominal x-ray showing dilated intestinal loops.

It was decided to perform a colonic opaque enema, in which the rectum and sigmoid showed signs of caliber reduction in the transition at the sigmoid level (figure 2).

**Figure 2:** Colonic opaque enema showing rectum and sigmoid with signs of decreased caliber, at the level of sigmoid transition.
A biopsy was indicated with a histological study showing the absence of ganglion cells in the distal, proximal and sigmoidal intestine. The most frequent manifestations of enterocolitis consist of abdominal dilation, constipation, vomiting and delayed meconium elimination. [10,11,17,18] In one study, neonates had 90% abdominal distension, 67.1% vomiting, 23% of constipation and 51% of cases, with delay in the elimination of meconium (> 48h 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 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, there is a predominance of constipation, occurring in 13 to 69% of cases [2]. Many authors defend the use of preventive measures that include the routine use of rectal irrigations in the postoperative period, long-term administration of metronidazole orally, use of probiotic therapy and diversion of enterostomy [18,19].

A simple abdominal radiograph may show an enlarged small intestine or proximal colon. Tests available for diagnosis include contrast enema, anorectal manometry, full-thickness rectal biopsy and rectal suction biopsy [9,14].

Treatment is done through a surgical procedure in which it aims to remove the aganglionic segment and reconstruct the intestinal transit. The most used surgical techniques are: Soave and Duhamel. [20-24] The first one is based on a colectomy and mucosectomy of the rectum with the accomplishment of a colon-anal tremo-terminal anastomosis with the normoganglionic segment. The second technique, on the other hand, consists of a colectomy with Hartmann’s bag and a termino-lateral colorectal anastomosis on the posterior wall of the rectum [1]. Both techniques create a reservoir for feces to be collected similarly to a native rectum after enterectomy [20-22].

After surgical treatment, a low cure rate of less than 20% was observed. Most of the children who are followed up on an outpatient basis complain of fecal incontinence [2].

Patients with a ganglion segment smaller than 20 to 40 centimeters became more likely to use Total Parenteral Nutrition (TPN) due to the small part of the absorptive intestinal surface after removal of the affected segment. The only way to avoid TPN is intestinal transplantation, aiming to improve the quality of life and decrease morbidity and mortality [7]. It is noted that even if the entire affected segment can be removed, there is no complete recovery of the patients, thus, it becomes 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, we aim to emphasize the need to alert or request a better understanding of the clinical picture of HD, as well as its complications and the importance of making an early diagnosis. In addition to raising awareness among emergency doctors as graves that cause this comorbidity, which may occur after the surgical procedure.

**References**

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