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Hirschsprung's Disease in Adults, A Case Report

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Abstract **Case Report**

Hirschsprung's disease (HD) is a rare congenital disease characterized by the absence of ganglion cells in the distal rectum and which is usually diagnosed in early childhood. We speak of Hirschsprung's disease of adults in patients where the diagnosis was made after the age of 10 years, it is a rare but not exceptional condition. This should be borne in mind in young adults with a history of chronic constipation. Due to the rarity of Hirschsprung's disease in adults, patients are rarely referred for evaluation, including rectal biopsy, further delaying diagnosis. The elements of the diagnosis are primarily clinical. The basic treatment for Hirschsprung's disease recognized in adulthood is surgical resection of the lymph node segment of the intestine with the intention of creating an anastomosis with healthy and properly innervated tissue. In this regard, we present the case of a 22-year-old young man, followed in the gastro-entero-hepatology department at the Arrazi hospital of the Mohammed VI University Hospital in Marrakech for aetiological assessment of a chronic constipation of terminal appearance, complicated by 2 episodes of sigmoid volvulus resolved by rectal probe detorsion, the rectal biopsy of which revealed aganglion in favor of Hirschsprung's disease.

Keywords: Adult Hirschsprung's disease - Chronic constipation - sub-occlusive episodes - area of aganglion - surgical resection.

INTRODUCTION

Hirschsprung's disease (HD) is rare congenital disorders in which ganglion cells are absent in the distal rectum and is typically diagnosed in infancy. Patients with a diagnosis of HD in adulthood commonly present with a long-standing history of constipation with attempted management with laxatives, motility agents, irrigation, or manual disimpaction [1]. Given the rarity of adult HD, patients are infrequently referred for evaluation, including rectal biopsy, which further delays diagnosis [2, 3]. Definitive treatment for HD includes resection of the aganglionic segment of the bowel [4] with the intention of creating an anastomosis to healthy, properly innervated tissue [5]. In some cases, diversion of the fecal stream may be required. Surgical approaches for the treatment of HD include procedures ranging from a three-stage approach to a minimally invasive one-stage approach [3]. However, these treatments are primarily studied in the treatment of infants with HD and little is known about the treatment and postoperative outcomes for adults with HD aside from case reports in the literature. This study aims to utilize a national database to identify the surgical treatment of Hirschsprung's disease and describe postoperative outcomes in the adult population. In this regard, we report the case of a 22-year-old patient, followed in the gastroentero-hepatology department at the Arrazi hospital of the Mohammed VI University Hospital of Marrakech for etiological assessment of a chronic constipation of terminal appearance, complicated by 2 subocclusive episodes, then operated for a dolichosigmoid with segmental resection of the sigmoid colon (26cm) with colosigmoid anastomosis, having benefited from a rectal biopsy which revealed aganglion in favor of Hirschsprung's disease.

OBSERVATION

We report the case of a 22-year-old patient who was admitted to the gastroentero-hepatology department at the Arrazi hospital of the Mohammed VI University Hospital of Marrakech for etiological assessment of a chronic constipation of terminal, intermittent appearance, at the rate of a normal bowel movement by 2 to 3 weeks, associated with asymmetric abdominal distension with atypical abdominal pain. Complicated by 2 episodes of sigmoid volvulus

resolved by untwist by rectal probe, then operated for a dolichosigmoid with segmental resection of the sigmoid colon (26cm) with colosigmoid anastomosis. Examination of the abdomen noted a slightly distended abdomen with tenderness of the left flank, so inspection of the anal margin and digital rectal examination were unremarkable. Imaging was carried out in particular an ASP: colonic distension predominant on the left with absence of rectal ventilation, supplemented by an abdominal CT (Figure 1): Aeric distension of the sigmoid colon and the transverse colon upstream of a fecal impaction of the upper rectum and the rectosigmoid hinge. Anorectal manometry was performed finding an RRAI present, incomplete relaxation of the anal canal on exertion. A surgical rectal biopsy was performed with the pathological study: hyperplasia of the nerve filaments in the absence of nerve ganglion cells. The diagnosis of Hirschsprung's disease was retained and the patient was proposed for surgical resection of the pathological aganglionic segment and with restoration of digestive continuity while respecting the sphincter system.

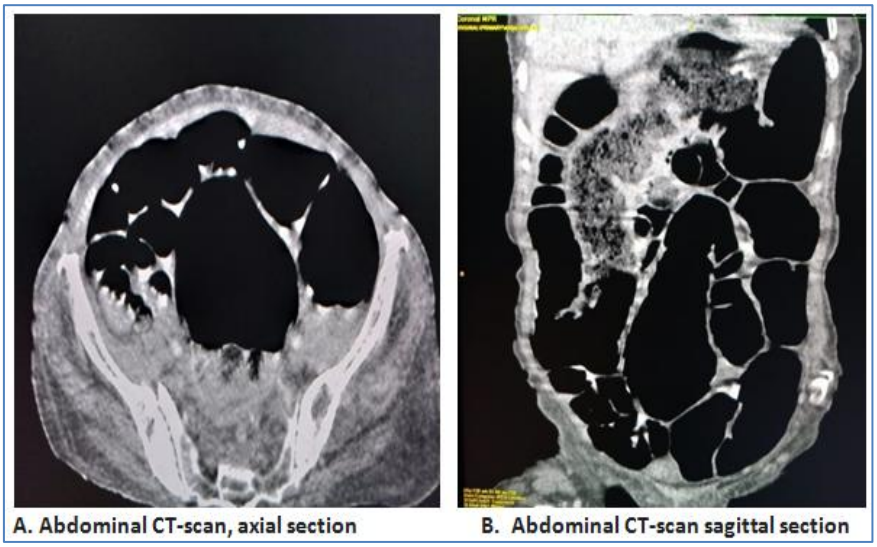

Fig-1: Aeric distension of the sigmoid colon and the transverse colon upstream of a fecal impaction of the upper rectum and the rectosigmoid hinge

DISCUSSION

Hirschsprung's disease (HD) is a rare condition with an incidence of around 1 / 5,000 births. It is usually diagnosed in the neonatal period; it is the most common cause of obstruction in children. It is caused by incomplete migration of neural crest cells during embryonic development, resulting in a lack of ganglion cells in the distal colon [6]. This aganglionic segment contracts, creating an obstruction and dilation of the proximal colon, which is normally innervated.

Patients generally present in the neonatal period with abdominal distension, bilious vomiting, enterocolitis, delayed meconium or stool emission, and growth retardation [7]. The average age at diagnosis is 2.6 months, but patients with less severe disease may not be diagnosed until later in childhood or even into adulthood [8]. A review via PubMed of the English-language literature over the past 10 years showed that there have been approximately 34 published cases of adults with untreated Hirschsprung's disease, many of whom experienced acute colonic perforation or a sigmoid volvulus. Diagnosis is most accurate with rectal aspiration biopsy, but abdominal imaging, barium enema, and anorectal manometry may also be helpful in the diagnosis [8, 9].

We speak of adult HD in patients diagnosed after the age of 10 years [10]. The actual incidence of this clinical form is unknown but has been estimated at 2% [11]. Almost 550 cases of Hirschsprung's disease in adults have been traced in the English literature since 1950 [12]. Authors [10] have reported cases of Hirschsprung's disease in patients over 30 years of age without a history of constipation or other symptoms during their childhood. This can be explained by very attentive maternal care made mainly of regular evacuating enemas which allow these patients to reach adulthood before the surgical indication is asked. Regarding sex, just like the form of the newborn, there is a clear male predominance of the adult form of Hirschsprung disease with a male / female sex ratio of 4.1 with an average age of 24.1 years [13]. Clinically, a history of chronic constipation evolving since the neonatal period has been found in most patients in the various publications [14, 15]. Usually, it is a major constipation which necessitated recourse to laxatives and often to enemas, the evolution of which may have been interrupted by unexplained improvement or on the contrary true occlusion which may have led to laparotomy or even colectomy [16]. Acute bowel obstruction was the reason for consultation in 1.8% of cases in the Doodnath meta-analysis [12]. Usually it is a low occlusion secondary to a fecal impaction. However,

it was secondary to a sigmoid volvulus. Alagumuthu [17] reported 3 new cases of HD in adults revealed by sigmoid colon volvulus. Abdominal distension secondary to fecal impaction, when it is important and when it is not taken care of quickly, could have urinary complications such as acute urinary retention, itself could be indicative of HD as in the case of Loganathan [18] published in 2013, or thoracic and respiratory complications such as mediastinal deviation, pulmonary atelectasis or pneumonia [19].

For additional examinations: The abdomen without preparation: The only direct sign of the affection on the X-ray is the visibility of a small caliber rectum. This aspect is rarely observed, the rectum is often empty of air. The more frequent indirect signs are those of an obstructive syndrome: presence of meconium or stercoral residues, distension of the small intestine and colon. Signs of enterocolitis or pneumoperitoneum are rare, intraluminal calcifications exceptional. Barium enema: The transition zone gives a characteristic funnel-shaped image [20]. However, this zone may be absent in ultra-short forms (less than 5 cm) of Hirschsprung's disease [19]. The second sign, after the caliber jump sign, is the retention of the contrast medium seen in late images. The abdomino-pelvic scanner makes it possible on the one hand to essentially eliminate a tumor cause more frequent at this age and on the other hand to detect the radiological transition zone which corresponds to the level of aganglionia. Anorectal manometry: is a dynamic examination that allows the recording of pressures in the rectum, internal sphincter and external sphincter. In HD, this examination shows the absence of an inhibitory recto-anal reflex (RRAI) and an increase in the basal pressure of the internal sphincter. It is a nonspecific test for the diagnosis of HD: The inhibitory recto-anal reflex is absent in idiopathic megacolon and it can be present in the ultra-short forms of aganglionia limited to the anal canal [21], therefore the Manometry has a specificity of 95%, but its sensitivity is only 25%, potentially resulting in false negatives [22].

The biopsy: the surgical biopsy makes it possible to make the diagnosis of hypoganglionosis and intestinal neuronal dysplasia, the clinical and histological characteristics of which are similar to those of HD. Ten out of eleven patients (90.9% of cases) in Duncan's series [23] had the diagnosis of Hirschsprung's disease confirmed through surgical biopsies. Aspiration biopsy provides an inadequate specimen with intractable results in one in three patients, they concluded that this technique gives the best results when applied in subjects under 3 years old [24].

There is no consensus on the actual length of the physiological hypoganglionosis segment in adults or on the required level of biopsy. A study carried out by Ricciardi [25] on 35 cadavers concluded that the average length of this zone is 24.4 ± 10.9 mm (distance calculated on the posterior wall of the rectum from the pectinate line) with extremes of 7.5 and 50 mm. A distance of 6 cm is retained by the majority of authors as sufficient to detect the area of aganglion during biopsies.

Treatment: The principle of surgical treatment for Hirschsprung's disease consists of resecting or excluding the diseased part of the digestive tract (area of aganglion) while preserving the innervation and function of the sphincter system [26].

Choice of technique: Fairgrieve [27], in 1963 described a first therapeutic approach for the management of Hirschsprung's disease in adults. This approach was based on a two-stage surgery: the first stage consisted of performing a colostomy in order to prepare the colon by evacuating the fecaloma and the second stage was devoted to surgical resection and colo-anal anastomosis a few months later. The author defended his strategy by the time required for the initially dilated colon to regain its usual dimensions after colonic bypass, and to be able to make an anastomosis outside of an incongruity between two segments of the digestive tract.

A second therapeutic strategy bringing patients to the operating room only once has been adopted by several authors. This is the case of Vorobyov [28] who opted for this approach in 67.8% of the cases of his patients. The two-step approach is reserved, according to these authors, for patients initially operated on as an emergency for acute intestinal obstruction or peritonitis by colonic perforation.

CONCLUSION

Hirschsprung disease in adults is a rare but not exceptional condition. This should be borne in mind in young adults with a history of chronic constipation. The elements of the diagnosis are primarily clinical. Anorectal manometry is a non-specific examination for the diagnosis of Hirschsprung’s disease, surgical biopsy in adults can confirm the diagnosis. The basic treatment for Hirschsprung's disease recognized in adulthood is surgical resection, although the choice of technique depends mainly on surgical expertise. Long-term functional results are usually satisfactory.

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