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Radiology

Hirschsprung's Disease in Adult Revealed by an Occlusive Syndrome: A Case Report

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Abstract **Case Report**

Hirschsprung's disease is a rare congenital disorder that primarily affects the rectosigmoid or rectal segments of the large intestine, resulting in abnormal motor function, persistent contraction of the aganglionic segment, and eventual dilation (mega-colon). While typically diagnosed in infancy or early childhood, it can also occur in adults and can be challenging to diagnose due to its rarity and production of mild symptoms in early stages. We present a case of an adult patient who was diagnosed with HD after experiencing constipation for 2 decades, leading to an occlusive syndrome. Clinical, radiographic, and computed tomography findings play a crucial role in diagnosing and managing this rare disorder in adults. Surgical resection of the aganglionic segment is the mainstay of treatment, and early intervention is crucial to prevent complications.

Keywords: Hirschsprung's Disease - Occlusive Syndrome – Adult – CT.

INTRODUCTION

Hirschsprung's disease (HD) is a congenital disorder caused by disrupted embryological development of the large intestine, resulting in the absence of submucosal and myenteric neural plexuses, primarily affecting the rectosigmoid or rectal segments (Fu *et al.*, 1996). Although most cases are diagnosed in infancy or early childhood, HD can also occur in adults, albeit infrequently.

In this report, we describe a case of an adult patient who was diagnosed with HD after experiencing constipation for 2 decades, leading to an occlusive syndrome. Clinical, radiographic, and computed tomography findings play a crucial role in diagnosing and managing this rare disorder in adults.

CASE REPORT

We present a case of a 25-year-old male patient with a history of chronic constipation since childhood. The patient underwent surgery in 2017 for an abdominal surgical etiology of unknown cause. He presented to the department with a sub-occlusive syndrome, characterized by diffuse abdominal pain, cessation of bowel movements with preserved gas emission.

During the physical examination, the patient was conscious and hemodynamically stable, with a distended abdomen and abdominal tenderness upon palpation. Rectal examination revealed an empty rectal cavity. Abdominal X-ray imaging showed significant fecal stasis throughout the colon with some peripheral hydro-aerial levels.

To investigate the etiology of fecal stasis, abdominal and pelvic computed tomography was performed. The results revealed enormous distension of the colonic frame and sigmoid, with a maximum estimated diameter of 14cm at the sigmoid colon **(Fig 1-Fig 2)**, site of significant fecal stasis without clearly detectable caliber disparity or hydro-aerial levels, with thinning of the wall in some places and loss of individualization of the colonic haustrations **(Figure 3)**. However, the underlying rectum was collapsed and appeared to be of normal caliber **(Figure 4-5)**.

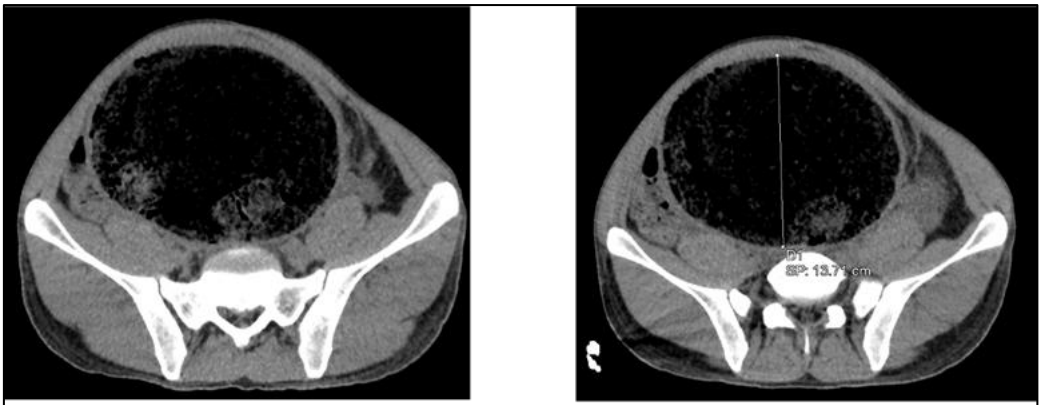

Fig 1 -2: CT scan without injection, axial section showing dilatation of the sigmoid to 14 cm

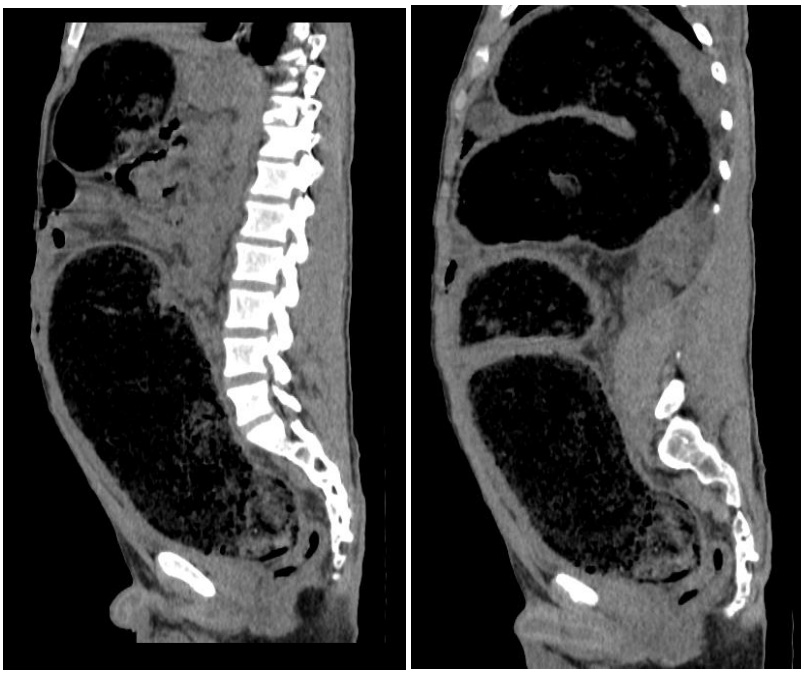

Figure 3: CT scan without injection, coronal section a dilatation of the colonic framework with absence of individualization of the colonic haustrations, without detectable zone of disparity of the caliber

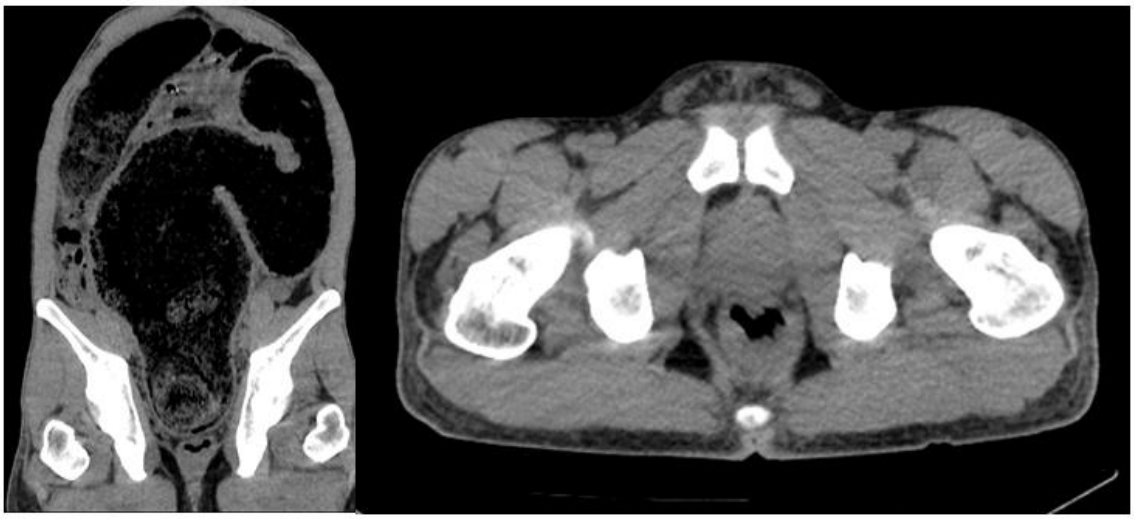

Figure 4 -5: CT scan without injection, axial and coronal sections, showing sigmoid dilatation with empty rectal ampulla without parietal thickening antero-posterior diameter.

The dilatation of the colonic frame was responsible for compression of the bladder with moderate upstream ureterohydronephrosis.

In the context of an occlusive presentation and increased risk of perforation, the initial emergency intervention consisted of establishing a decompression colostomy with colonic drainage, as well as a rectal biopsy to confirm aganglionosis **(Fig 7)**. The histopathological examination confirmed Hirschsprung's disease, with diffuse aganglionosis. Subsequent management involved surgical resection of the aganglionic segment, resulting in restoration of colonic continuity.

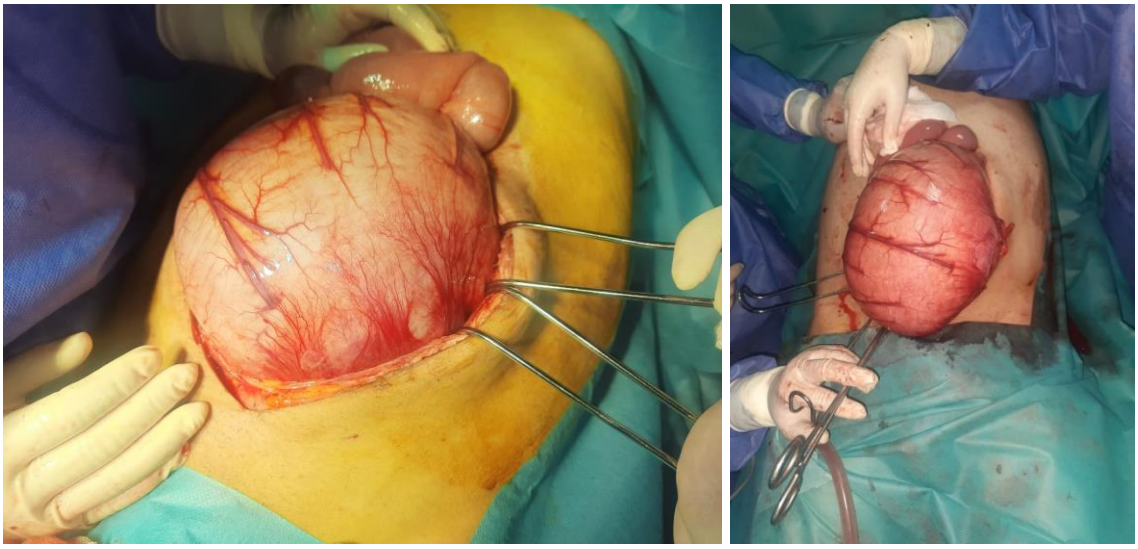

Figure 7: Pictures of the operating room showing the major dilatation of the sigmoid colon

DISCUSSION

Hirschsprung's disease (HD) is a rare condition affecting approximately 1 in 5000 live births and is typically diagnosed during the neonatal period. (Qiu *et al.*, s. d.) However, a small number of cases may remain undiagnosed until after 5 years of age, and its incidence in adults is unknown as this diagnosis is often overlooked in this population (Kim *et al.*, 2008).

The primary defect in HD involves the absence of ganglion cells in the Meissner and Auerbach plexuses of the affected segment of the large bowel, .This leads to abnormal motor function of the colon, persistent contraction of the aganglionic segment and eventual dilatation (mega-colon) and perforation of the normally innervated colon (Martucciello, 2008). Symptoms include refractory constipation, abdominal distension and pain, and palpable fecal mass(Vorobyov *et al.*, 2010).

Diagnosis in adults can be challenging due to the rarity of the condition and the production of mild symptoms in early stages of the disease(Vorobyov *et al.*, 2010). Adult Hirschsprung's disease should be suspected when there is a long history of constipation from early childhood, associated with abdominal distension and colic. Other symptoms include anorexia, vomiting, malaise, lethargy, weight loss, borborygmi and respiratory or cardiac embarrassment with gross distension(Goto *et al.*, 1984).

Conventional radiography shows colonic dilatation with stercoral retention or gaseous distension, and CT scans are useful for anatomic localisation, showing large bowel dilatation among the transition area, and excluding other causes of chronic constipation and megacolon in adults, such as colorectal cancer and volvulus(Kim *et al.*, 2008).

Our case presented a long history of constipation with surgical resection at the age of 17, but without documentation, which could be related to Hirschsprung's disease but was underdiagnosed. The preservation of gas emission made the picture of obstruction incomplete, which was confirmed by CT scan later on, with the enormous distension of the colonic frame and the absence of a zone of caliber disparity. The existence of a normal zone at the rectal level and the absence of wall thickening or tumor lesion at this level led us to consider aganglionosis despite its rarity at this age. The absence of signs of digestive complications and the history of frequent recurring constipation support this diagnostic hypothesis.

Anorectal manometry typically demonstrates no internal anal sphincter relaxation in response to rectal distension. Diagnosis is confirmed by rectal biopsy from the narrowed segment(Wheatley *et al.*, s. d.).

Surgical resection of the aganglionic segment is the mainstay of treatment, and early intervention is crucial to prevent complications. The objective of

treatment is to restore continuity between the two healthy segments (Martucciello, 2008).

CONCLUSION

HD in adults is rare. It should be considered in front of any history of chronic constipation refractory to treatment since childhood. The diagnosis is based on a combination of clinical, manometric, radiological, and histological evidence. As in our case, it may rarely occur as massive colonic occlusion with risk of perforation, which requires emergency surgery.

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