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Adulthood presentation of Hirschsprung's disease

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ABSTRACT

In 90% of cases, Hirschsprung's disease (HD) in adults occurs before the age of five. The primary characteristic is the absence of ganglion cells in the colorectal segment, resulting in upstream colonic dilatation and functional obstruction. Any history of chronic constipation should raise suspicions of HD. A combination of radiological, histological, clinical, and manometric results forms the basis of the diagnosis. The mainstay of treatment is surgery, which involves cutting off the aganglionic segment and then re-establishing continuity between the two healthy segments.

Keywords: *Constipation; adults; Hirschsprung's disease (HD)*

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Introduction

A malformation of the large intestine's (colon's) embryonic development is known as Hirschsprung's disease (HD). It is characterised by an abnormality of the colon's motor function. This is because the ganglion cells of Auerbach's plexus in the muscularis and Meissner's plexus in the submucosa are absent [1]. In 90% of cases, HD's clinical symptoms generally appear before the age of five [2]. This causes an upstream dilatation of the colon and functional obstruction [3]. Surgery is a unique method of treatment.

Case Presentation

Case (1): A male individual aged 23 who has a history of altered bowel habitsfour months before surgery presented to the gastroenterology clinic with abdominal distension, no vomiting
O/E vitally stable
Abdominal examination revealed distension all over the abdomen.
Bowel sound +ve.
PR exam empty rectum.
Plain X-ray of abdomen: distension of the large bowel.
U/S revealed normal apart from considerable bowel distension.
CT scan of the native varietyshowed a hugely distended large bowel till the upper rectum (Fig. 1)

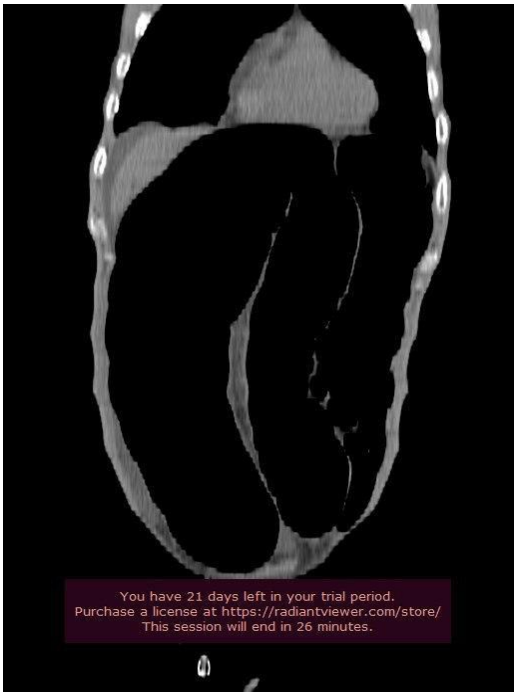

Figure (1): Hugely distended large bowel

Contrast enema showed a dilated large bowel with a narrowed segment between the junction of the upper and middle thirds of the rectum (Figure 2).

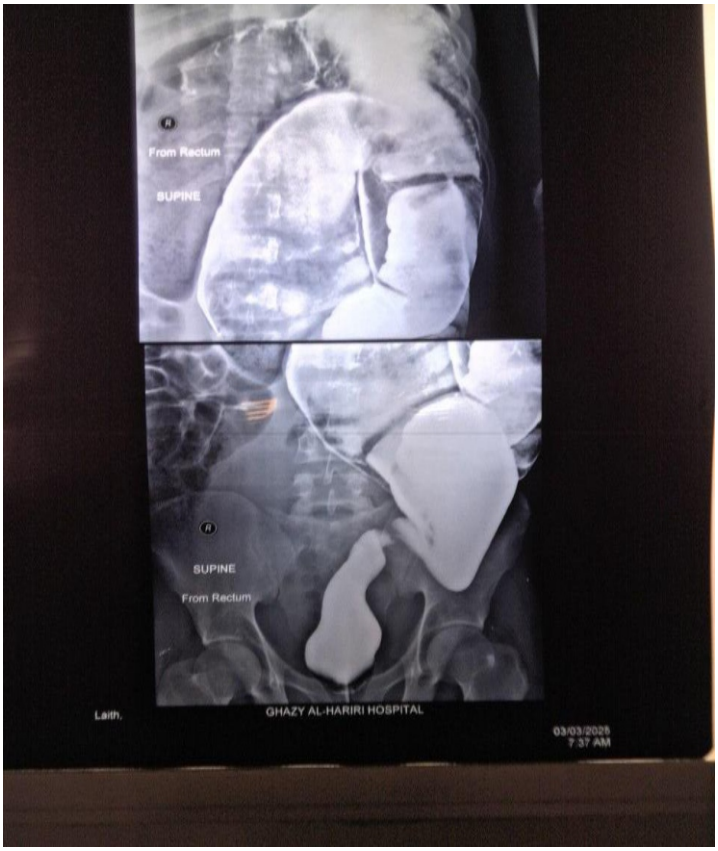

Figure (2): The Arrow showsthe narrowed segment.

A ccolonoscopy 10 cm from the anal verge cannot be passed with a badly prepared bowel.

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All the above clinical and radiological parameters necessitated the patient to be explored.
Laparotomy done through midline incision showed a hugely dilated large bowel from the ileocecal region till the junction of the upper and middle rectum.

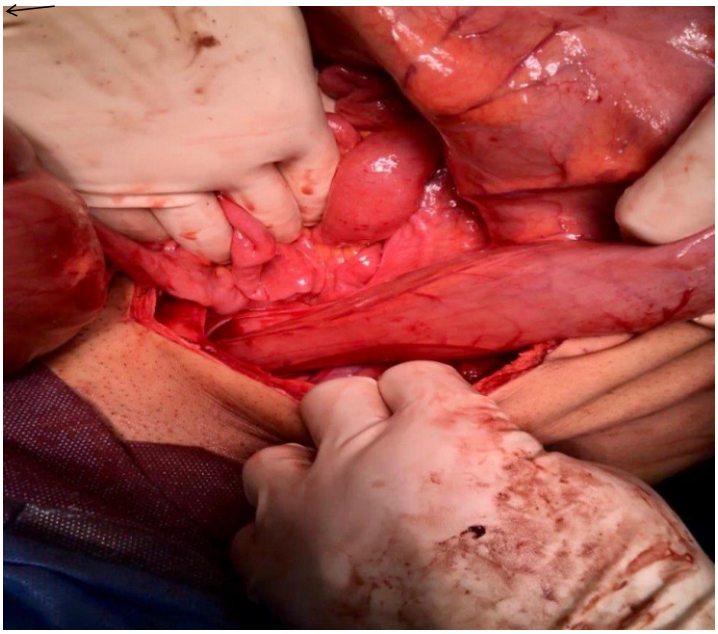

Figure (3): Narrowed segment between the upper and middle rectum.

Bowel as a reservoir of faecal material without any peristalsis, so decided to do total colectomy with ileo-rectal anastomosis and protective ileostomy as shownin Figure-4.
Specimens were sent for histopathology,the result was absent ganglionic cells in the colon, hypertrophied nerve trunk in the stenotic area in the rectum, just above the resection margin.
The combination of clinical, radiological, and pathological results favoursHirschsprung's disease.

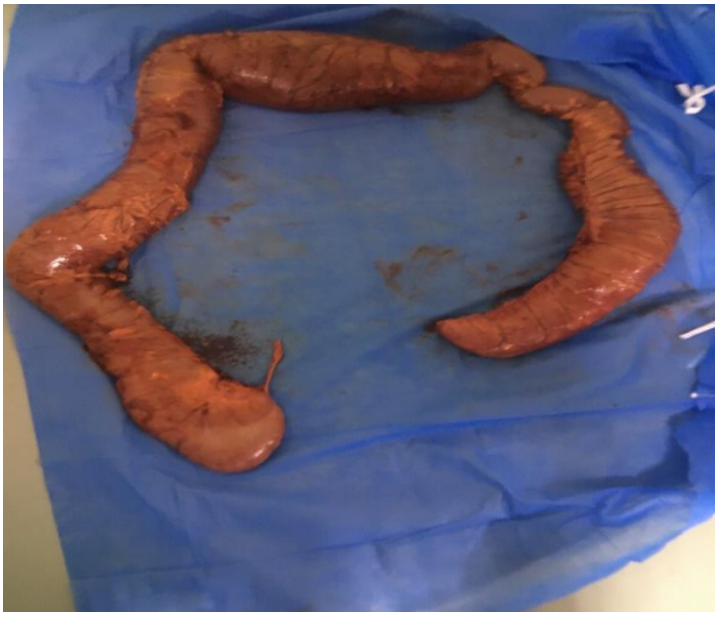

Figure (4): specimen of total colectomy with resection of the upper part of the rectum

Case (2)

A 25-year-old female with a history of absolute constipation of 10 days duration and lower abdominal pain, no vomiting, treated conservatively with analgesia and laxatives, but no benefit.
O/E vitally stable
Abdominal examination revealed lower abdominal distension and tenderness, mainly in the left lower abdomen.
Laboratory investigations were routine. Imaging: Plain X-ray of abdomen, distended large bowel mainly lower abdomen
CT of the native variety showed a distended segment of the large bowel with evidence of proximal and distal end stenosis of the dilated bowel (Fig. 5).

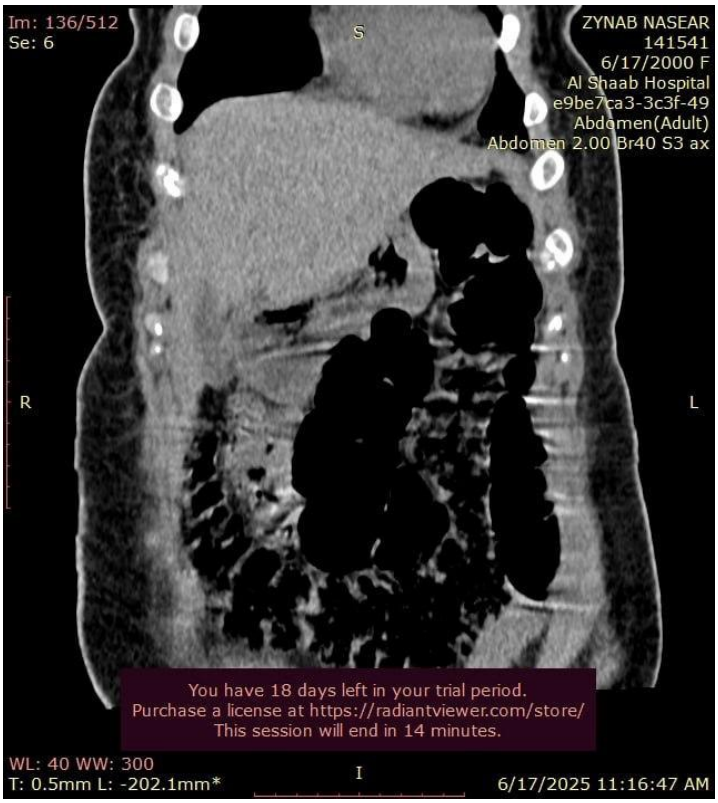

Figure (5): dilated large bowel with proximal and distal stenosis

Laparotomy done through midline incision revealed dilated sigmoid with stenosis of proximal and distal ends,no apparent mass intramural, intraluminal or band extramural,so resection of sigmoid colon with Hartmann procedure was done.
Biopsy result: absence of nerve plexus in submucosal and myenteric ganglion cells with nerve endings, favouring Hirschsprung's disease.

Discussion

About 1 in 5000 babies are born with HD. 600 cases have been diagnosed in adulthood [3–4], with only a small number being diagnosed after the age of five [2]. Many authors concur that patients aged 10 and up are diagnosed with "adult Hirschsprung's disease."
Adult patients are typically between the ages of 10 and 73, and the incidence of HD rises before the age of 30 [3]. A defect in the craniocaudal migration of neuroblasts originating from the neural crest, which starts at four weeks of gestation and ends at week seven with the arrival of neural crest-derived cells at the distal end of the colon, is the most widely accepted explanation for the cause of HD.When cells are unable to reach the distal colon, the segment becomes a ganglion and exhibits aberrant motor function, leading to HD [5].
Age has an impact on the clinical presentation. Abdominal distension, delayed passage of meconium, and bilious emesis are the main symptoms in newborns [6]. Adult HD is difficult to diagnose because of the short or ultra-short ganglionic segment, which may account for the delayed onset of symptoms, which are typically mild [7].

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A history is a crucial diagnostic tool. It displays symptoms like persistent constipation and stomach pain. Knowing the type and duration of constipation is part of history.

History should be employed for evaluating secondary causes of constipation. In order to rule out structural bowel changes or organic diseases, a recent and persistent change in bowel habits should be evaluated if it is not linked to a clearly identifiable cause of constipation (such as medications). This is especially crucial for older adults who also have anaemia or occult gastrointestinal bleeding, or who complain of excessive straining or a feeling of incomplete evacuation [8]. Colonic dilatation and a narrowing image that matches the ganglionic zone are demonstrated by barium enema and CT scan, which also rule out other causes of persistent constipation [9–10]. The anorectal inhibitory reflex is absent, according to anorectal manometry [7–11].

As the initial diagnostic measure, we recommend anorectal manometry and contrast enema instead of suction biopsy. Assume that a distinct transition zone is visible on the barium enema and that the manometry results are suggestive of HD. In that instance, the study is essentially pathognomonic of HD and aids in the planning of the surgical strategy; however, the results of a suction biopsy must be obtained to confirm it. In the event that manometry results are negative, a histological examination of surgical specimens is imperative. If there are no ganglion cells in the biopsy, HD is diagnosed. Hypertrophic nerve fibres, elevated acetylcholinesterase activity or staining in the muscularis mucosae, and reduced or absent calretinin-immunoreactive fibres in the lamina propria are examples of supporting findings [5].

Given the potential for complications, the primary treatment option for HD is surgery, which ought to be explored as soon as feasible [10]. A ganglionic segment is surgically removed, and the continuity between the two healthy segments is then restored [5]. The Swenson, Duhamel, and Soave procedures are therefore the three primary methods. The Duhamel technique [11] does not require rectum transaction [12–13] and consists of a retro-rectal trans-epiploic trans-anal pull-through. Typically, it is carried out in two phases [14]. In the first method, the rectum is sectioned from above the ganglionic zone, and then the upstream colon is lowered using a transanal technique. A transanal low colorectal anastomosis is the foundation of the second stage [10].

In order determine the severity of ganglionated bowel, additional mucosal biopsies are taken along the sigmoid's antimesenteric border as part of the Swenson [15] procedure. The rectum is dissected at the wall's contact because of the release of the pathological colon. This level involves a sigmoid rectal resection, an eversion of the rectal stump, a coloanal anastomosis, and a stapling procedure (lower than the Duhamel technique). Similar to Duhamel's technique, the Soave procedure [16] involves stripping the rectal mucosa while maintaining the rectum's muscular cuff; a coloanal anastomosis is carried out following the removal of a ganglionic segment of the colon [13]. The transrectal Swenson and Soave techniques necessitate a temporary upstream stoma. Some authors claim that by taking into account a less invasive method that involves laparoscopic sigmoid rectal resection followed by a trans anal lowering of the healthy colon without a stoma, surgery can have positive and fulfilling results.

When compared with the processes mentioned above, this method yields a more aesthetically pleasing and useful outcome [17].

The emergency nature of the situations and the uncertainty surrounding the diagnosis made the therapeutic decisions difficult for our patients. As a result, we decided on a total colectomy, a latero-terminal ileorectal anastomosis, and a Hartmann's procedure. The surgical specimen's histological results supported the HD diagnosis.

Conclusions: Adult HD is uncommon. When there is a history of persistent constipation, it should be taken into consideration. A combination of radiological, histological, clinical, and manometric evidence support the diagnosis. Histologically, it is identified by the lack of ganglion cells in a particular colon segment.

A history is a crucial diagnostic tool. Knowing the type and duration of constipation is an important part of history. Finding secondary causes of constipation should be another goal of history. If a recent and ongoing change in bowel habits is not linked to a clearly identifiable cause of constipation, it should lead to an evaluation to rule out organic diseases or structural bowel changes. HD is primarily treated surgically, though diagnostic procedures may also be carried out. It entails cutting off a ganglionic segment and then re-establishing continuity between the two healthy segments.

Additional facts: Individual participants. Every participant in this study gave their consent or waived it.

Conflicts of interest: All authors affirm the following in accordance with the ICMJE uniform disclosure form: Details of payment and services: Every author has stated that no organisation provided financial support for the work that was submitted.

Financial ties: All authors have stated that they have no financial ties to any organisations that might be interested in the work they submitted, either now or in the last three years.

Other relationships: Each author affirms that none of their other relationships or activities could be interpreted as having an impact on the work that was submitted.

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