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Adult Hirschsprung's Disease: A Rare Diagnosis

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Abstract

Hirschsprung's disease is a congenital abnormality in which there is absent neuronal ganglion cells in the submucosal (Meissner) and myenteric (Auerbach) neural plexuses mainly in the distal colon. Presentation is commonest in neonates and very rare in adults. We reported a case of adult Hirschsprungs disease in a young female patient who was successfully treated surgically. The high index of suspicion based on the patient's clinical features showed the increase awareness of Hirschsprung's disease as a differntial diagnosis of chronic constipation with chronic laxative use.

Introduction

Hirschsprung's Disease (HD), also called congenital aganglionic megacolon [1] is a rare motor disorder characterized by the total absence of intramural ganglion cells of the submucosal (Meissner) and myenteric (Auerbach) neural plexuses in the affected segment of the large bowel [2]. It occurs mostly the distal colon [3].

It is mostly diagnosed in neonatal period of life [4], especially in neonates who fail to pass meconium within 24-48 hours of birth [5]. It is relatively common, with some regional incidence ranging from 1:5000 - 1:7000 live births [6], but commoner in people of Asian descent [7]. There is a male preonderance especially in the commonest variant of recto-sigmoid disease [4]. Ninety-four percent of HD cases are diagnosed before the patient reaches 5 years of age, however, on rare occasion, mild cases of HD may go undiagnosed till adulthood [2].

Adult Hirschsprung's disease is very rare and frequently misdiagnosed as refractory constipation [2]. More than 600 adult cases have been reported in literature prior to now [8]. Presentation is usually that of constipation of long-standing duration with history of chronic laxative use.

Contrast enema is useful in establishing the diagnosis but full-thickness rectal biopsy remains the gold standard. The definitive treatment is surgical removal of the aganglionic bowel and to restore continuity of the normal bowel with the distal rectum, with or without an initial intestinal diversion.

A few studies have described adult HD in Nigeria [1,9,10] and this case will only add to the data suggesting that HD in adult, though uncommon but might not be very rare as currently thought.

Case report

An 18-year-old woman, a student of a tertiary instituition in Nigeria with a long-term history of constipation was referred to the gastroentrology clinic of a tertiary hospital in south-western Nigeria from a private clinic after unresponsiveness to the prescribed liquid parafin and high fiber dietary instruction. She presented with a 15-year history of constipation associated with chronic laxative use. She had tried several laxatives over the years but no permanent relief of symptoms. She had also practically changed her diet to that of mostly fluid in nature. In the last 3 weeks prior to presentation, she started having increasing abdominal distention with abdominal pain marked at the lower right and left region. She had no family history of similar complaints. She had no history suggestive of abdominal

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tuberculosis or metabolic disease. She also had no history of abdominal surgery. She didn't smoke nor take alcohol. She didn't use local herbs as medicinal remedies. Her genotype was AA and her blood group was O^+. Clinical examination showed the abdomen was grossly distended with visible peristalsis. There were no palpable masses and no organomegaly. Digital rectal examination revealed good anal sphincteric tone with a high rectal fecal load.

All of the routine laboratory investigations such as serum electrolyte/urea/creatinine, full blood count, blood glucose, liver function test, and urinalysis were all within normal limits. Colonoscopy done in the private clinic before presentation was reported as normal. Based on the above test results, a barium enema was ordered and it showed *luminal narrowing in the rectosigmoid junction with distention of the proximal large bowel loop while the rectum showed normal calibre (Figure 1).*

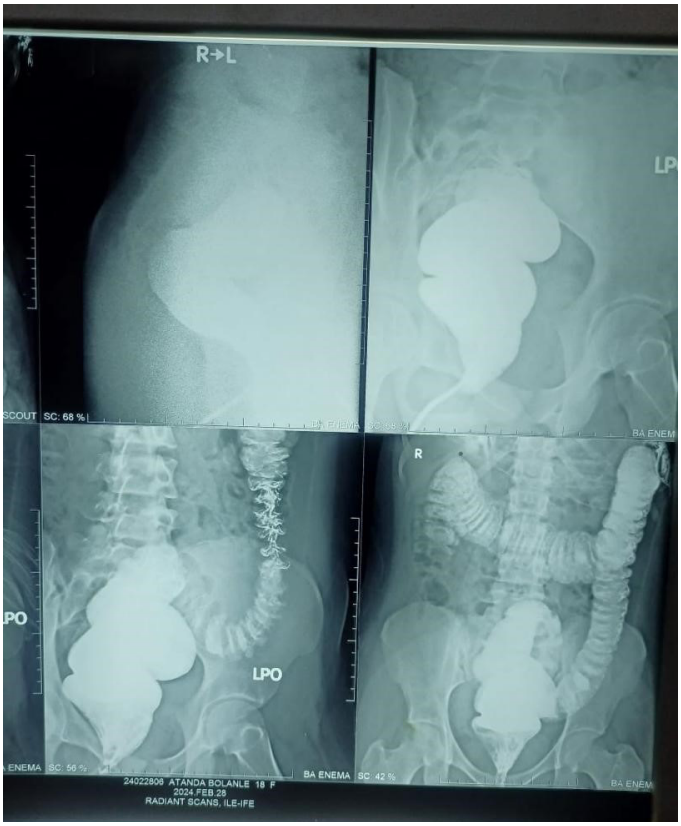
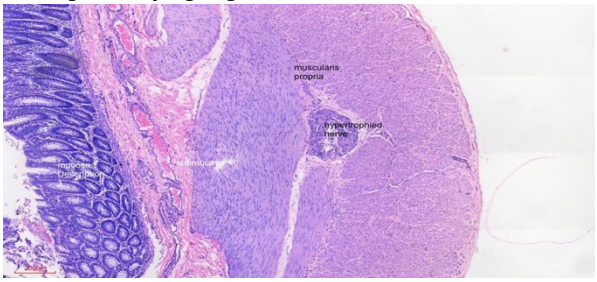


Image showed a focal persistent short segment (13 mm long) luminal narrowing demonstrated in the rectosigmoid junction. The proximal large bowel loops (including the descending colon, transverse colon, ascending colon) were grossly distended while the distal large bowel loop (rectum) showed normal calibre. No focus of contrast extralumination was seen (Figure 1).

This prompted the suggestion of a possible Hirschsprungs disease. A rectal biopsy which was the logical next step was done and it showed rectal tissue with an unremarkable columnar epithelium lining the mucosal surface*. There were no ganglion cells in the submucosa and muscularis propria. A pathological diagnosis of Aganglionosis was made*.



Section show Hematoxylin and Eosin stains of the colon showing muscularis propria displaying hyperplasia nerve bundle devoid of ganglion cell and muscular hypertrophy consistent with Hirschsprung disease (Figure 2).

The histology confirmed the diagnosis of HD, and she was subsequently referred for surgery. She had low anterior resection of the rectum plus stapled colorectal anastomosis and a protective transverse loop colostomy. The colostomy was later reversed 3 months after the initial surgery.

Patient is currently doing fine, and quality of life has greatly improved.

Discussion

Hirschsprung's disease is a congenital abnormality in which there is absent neuronal ganglion cells in the submucosal (Meissner) and myenteric (Auerbach) neural plexuses mainly in the distal colon [2,3]. It is mostly diagnosed in neonates but adult cases, though uncommon, have been reported. There are no data concerning the incidence or the prevalence of adult HD in nigeria except the few reported cases in literature.

Our patient is an 18-year-old woman who did not represent the common demographics seen in a disease that is commoner in children and male gender [4]. Her presentation of long-term constipation with chronic use of laxatives is typical of the few reported cases of adult HD. Her history of a progressive reduction in the response to laxatives over the years was also typical of most of the earlier reported cases in literature. Likewise, was her examination findings and investigations compared to earlier reported cases.

The diagnosis was made with a high index of suspicion based on the patients symtoms and history of self administered and prescibed medications despite the 15-year duration before presentation. This probably signifies increased awareness of HD in adults as a differential diagnosis of chronic constipation and excessive use of laxatives in the tertiary hospital setting but a persisting lack of awareness in lower levels of medical care which prevented the early recognition of HD in this patient. This allowed for the unneccassary use of laxatives and unwarrranted dietatry modifications over a long period of time by the patient. This is corroborated by the fact that most of the reported cases in Nigeria were made at the tertiary hospital [1,9,10].

The relative lack of serious complication such as Hirschsprungs Associated Enterocolitis (HAEC) might have been responsible for the uneventful surgical and post-surgical phase of the treatment of this patient. Presence of complications increases likehood of morbidity and mortality during surgery and post-surgical management[11]. The short segment aganglionosis type of disease in the patient might have been one of the strong reasons for the absence of serious complications [12].

In conclusion, adult HD should be considered as a differential diagnosis in patients, especially young adults, that present with chronic constipation and chronic and excessive laxative use. Prompt barium studies in such patients will be highly beneficial and rewarding.

Author declarations

Patient consent

The authors certify that they have obtained all appropriate informed patient consent. In the form, the patient gave her con-

sent for the clinical information and the included image to be reported in the journal. The patient understands that her name and initials will not be published, and due effort will be made to conceal her identity. However, anonymity cannot be guaranteed.

Conflicts of interest

The authors declare no conflicts of interest.

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