Residual Hirschsprung's Disease in Adulthood: Management Challenges at a Nigerian Tertiary Hospital - A Case Report

[1]*O.G. Habeeb, [1]I.O. Olaoye, [1]S. N. Irmiya, [1]S. O. Oyewale, [1]O. A. Jimoh, [2]Z. A. Suleiman

[1]Department of Surgery University of Ilorin Teaching Hospital, Ilorin Nigeria. [2]Department of Anesthesiology, Geisinger Medical Center, Geisinger Health System, Danville, Pennsylvania, USA.

Abstract

Background: Hirschsprung's disease (HD)is a developmental condition that is found mainly in the paediatric population. However, a few of them can still present to adult surgeons with a residual aganglionic intestinal segment despite an initial operative intervention. Challenges in management occur largely from the paucity of surgical experience when compared with the paediatric population.

Case Report: A 21-year-old man presented to our adult emergency room 13 years after a trans-anal pull-through with features of obstruction. The patient was diagnosed with residual HD. He underwent an initial colostomy followed by the Duhamel procedure which was complicated by anastomotic dehiscence. He subsequently had an ileostomy which was reversed after four months when there was no contrast extravasation at the site of anastomosis.

Conclusion and Recommendations: Here, we present our experience in the management of a patient with residual Hirschsprung's disease who was successfully managed with a two-stage approach after a failed single-stage attempt. This report highlights the challenges of incomplete resection of HD in the paediatric age group presenting to the general surgeon in adult life.

Keywords: Adult, Constipation, Residual lesion, Hirschsprung's disease, Challenges

Background

Since 1880, when the first report on Hirschsprung's disease (HD) was made, various treatment modalities have been explored for managing residual HD in adults. The selected treatment is dependent on the patient's presentation and the experience of the surgical team. [1]The routine use of frozen section biopsy in HD should prevent the retention of aganglionic intestinal segments. However, in the absence of intraoperative pathologic assessment, a small proportion of patients with HD will present in adulthood with persistent constipation after Transanal pull-through surgeries due to incomplete resections of the diseased intestinal segment. [2] Many babies with Hirschsprung disease will present within 48 hours of life with delays in passing meconium and other features of intestinal obstruction.[3]

Correspondence to:

Habeeb Olufemi Gbenga
Division of General Surgery, University of Ilorin Teaching Hospital
Email: habeeb.og@unilorin.edu.ng
ORCID: https://orcid.org/0000-0002-8844-708X

However, a proportion of these patients will have residual disease in adulthood despite initial operative interventions. Consequently, these patients may develop poor management outcomes due to increased morbidity and mortality. [4]

This case report describes the management of an adult who presented to general surgeons after a trans-rectal pull-through surgery. This highlights the possible challenges of managing such patients in a resource-poor setting. This case report was written in accordance with CARE guidelines (for CAse REports). [5]

Case Report

A 21-year-old adult male who presented to the emergency room with recurring constipation, persistent abdominal distension and occasional colicky abdominal pain for "*as long as he could remember*" (several years). He had undergone a Transanal pull-through procedure 13 years previously for a similar presentation. The surgery was uneventful. There was no family history of congenital disease. Examination revealed a tachycardic young adult with a grossly distended abdomen but with no features of peritonitis. Abdominal radiography (Figure 1) showed gross dilatation of the sigmoid colon and rectum, with

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intraluminal mottling. Barium enema (Figure 2) revealed a grossly distended recto-sigmoid segment with a demonstrable transition zone. Other laboratory test results were normal.

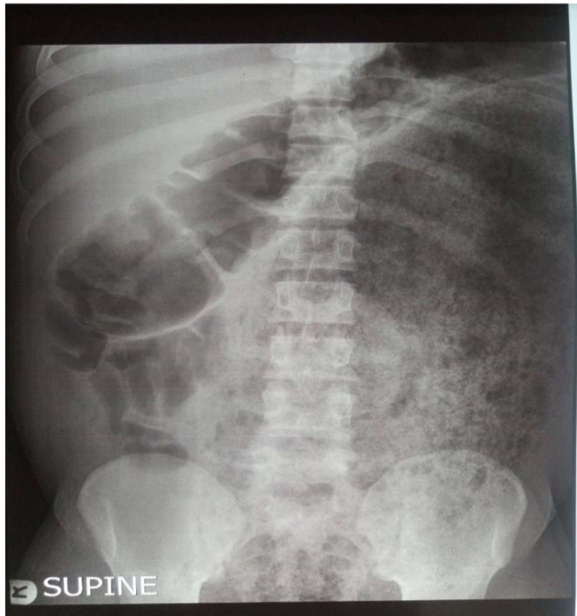

Fig 1: Abdominal plain radiograph (supine view)

A diagnosis of Residual HD was considered in this patient. He was resuscitated and had an exploratory laparotomy, with intraoperative findings of; grossly dilated distal sigmoid colon and narrow proximal rectum (Figure 3). Sigmoid colectomy with exteriorization of the remnant of the sigmoid colon and proximal rectum as divided colostomy was done. A full-thickness rectal biopsy and proximal sigmoid biopsy were performed, and histology confirmed aganglionosis in the rectum and the presence of ganglion in the proximal sigmoid colon, respectively.

He was nutritionally optimized, and 4 months later underwent resection of the diseased segment and a Duhamel procedure (this was the second surgery). However, the postoperative period was complicated by anastomotic leak with intra-abdominal collection necessitating re-operation and ileostomy (third surgery). He made remarkable improvements and was allowed home to fully recuperate.

A distal loopogram which was done about three months later, showed no contrast extravasation at the anastomosis. He then underwent ileostomy reversal to ensure bowel continuity (fourth surgery). He returned to his regular activities with a gradual return to normalcy in bowel function, and the last clinic visit revealed a complete recovery.

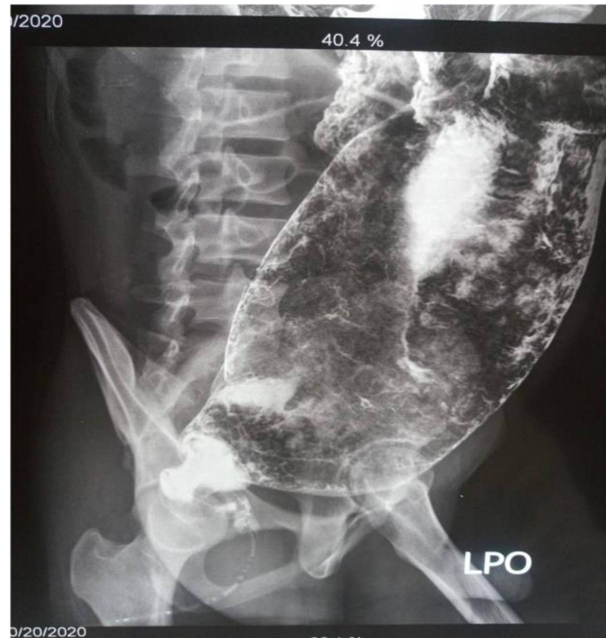

Figure 2: Barium Enema (left oblique posterior)

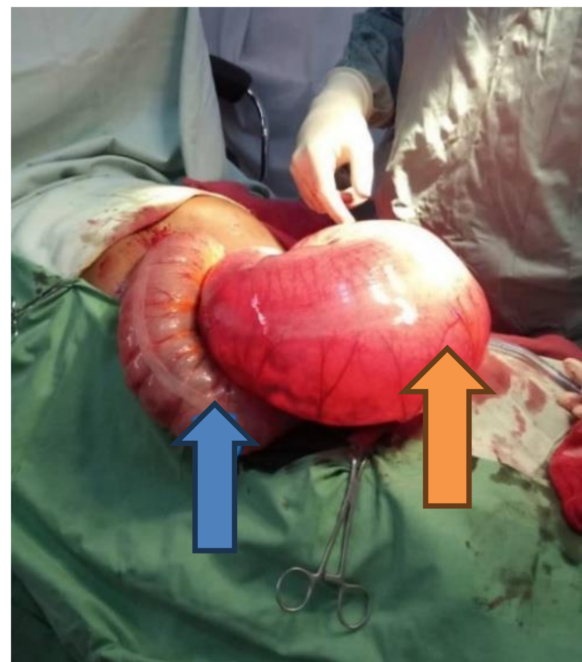

Figure 3: Blue Arrow (Transverse Colon), Orange Arrow (Sigmoid Colon)

Discussion

Although the results of pull-through for Hirschsprung disease have generally been regarded as favourable, a minority of these patients require reoperation because of dysfunctional stooling after pull-through surgery. Patients with dysfunctional post-pull-through stooling can be divided into those with obstructive symptoms (such as constipation or enterocolitis) and those with non-obstructive symptoms (such as soiling or incontinence). Presentation usually varies, as some may present acutely,

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as in our case, or may present with chronic symptoms. The various causes leading to obstructive type of dysfunctional pull-through include mechanical obstruction, retained or residual aganglionosis, and a transition zone in the pull-through.

According to a meta-analysis carried out by Friedmacher and Puri in 2011, residual aganglionosis and transition zone tissue accounted for persistent bowel symptoms in 33% of patients undergoing a second, corrective pull-through procedure.[6] In addition, a study conducted by Pena et al. revealed that out of 54 patients who presented for reoperation following an initial intervention, 8 had residual aganglionosis detected on rectal biopsy. [7] Patients presenting with post-pull-through complications should be thoroughly assessed by experienced surgeons in an institution that has the infrastructure to evaluate, diagnose, and treat these complex problems.

An algorithm should be developed to ensure that patients are thoroughly evaluated, which should include a comprehensive history and physical examination. Previous history of intestinal obstruction and surgery in childhood without a frozen section raised a suspicion of residual HD. It is important that a rectal examination be performed during physical examination in order to check for anal tone which may be reduced or absent in some of these patients. However, when a gush of gas or liquid stools occur after removal of examining fingers, this may suggest chronic obstructive symptoms or Hirschsprung associated enterocolitis (HAEC).

Other important aspects of evaluation include investigations such as imaging techniques. These include plain abdominal radiographs (erect and supine views), contrast enema, and computed tomography scans.
An upright plain abdominal film allows one to delineate the degree of obstruction and may also show a “cut-off” sign suggestive of active or chronic HAEC. A radiographic contrast enema can differentiate between anastomotic stenosis, stricture, or twist of the pull-through segment, as well as a chronically dilated rectosigmoid segment.

In addition, a rectal biopsy which could be either mucosal or submucosal or full-thickness, is an important and precise method for the evaluation of chronic problems after a pull-through. However, this diagnostic evaluation tool is met with a few challenges, for example, identification and distinction of the dentate line, anastomosis, and the distance between them may be difficult. This is because with growth, the distance from the dentate line to the anastomosis may become greater than expected, or it may lengthen what was a relatively short aganglionic segment at the time of the initial operation.

Other useful investigation modalities are anorectal manometry and flexible sigmoidoscopy, alongside blood investigations, such as full blood count, serum electrolyte, urea and creatinine, grouping, and cross blood matching of blood for repeat pull-through procedures. Adequate resuscitation of the patient is paramount, especially in cases of intestinal obstruction, where fluid and electrolyte derangements are common. After stabilization, definitive surgery can be embarked on. There are a couple of operative procedures that exist for redo pull-through in Hirschsprung’s disease, but it is important to note that these procedures are much more difficult to perform than the original operation. Soave, Swenson, or Duhamel could be performed during the reoperation. In a series by Mohammed Elsherbeny and Sameh Abdelhay, four patients had retained aganglionosis, and redo pull-through was required in all of them. The redo procedure was trans-anal endorectal pull-through in the two patients who underwent initial trans-anal endorectal pull-through and abdominal assisted pull-through in the two patients who underwent the initial Duhamel procedure. [8] Choice of Duhamel in this index case was based on Surgeon’s preference and expertise.

In this group of patients, there could be difficulty in recognizing the pattern of presentation among adult surgeons which could result in toxic mega colon, sigmoid volvulus, and bowel perforation. [9] Based on an organized algorithm of treatment and the nature of presentation of patients, repeated dilatations, use of botulinum toxin, and posterior myotomy or myomectomy can be used in cases of strictures and internal anal sphincter achalasia. However, if these measures fail, a repeat pull-through surgery should be performed.

Retained aganglionosis, especially if it exceeds a length of 3 cm, is an absolute indication for redo pull-through surgery, and a transitional zone pull-through with dilated neorectum is also considered an indication for surgery. Decompressive techniques such as colostomy can be used in the interim in patients who present acutely. The approach to performing a repeat pull-through varies and is dependent on the type of procedures previously performed. Laparoscopy can often be useful in exploration and obtaining levelling biopsies, especially in cases where the surgeon has expertise in minimally invasive surgeries. One of the advantages of the laparoscopic approach, aside from other benefits of minimal access surgery, is that it allows a deeper dissection into the pelvis.

If laparoscopy is not performed, a lower abdominal midline incision is ideal, or a Transanal approach may be used. Thorough lysis of adhesions is frequently required and should be performed. It is important to establish an appropriate level for the pull-through segment, and once that has been done, complete mobilization of the distal colon is performed. Two critical challenges at this point

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are to ascertain the vascularity of the bowel and to ensure adequate length for a tension-free anastomosis.
In patients with a previous colostomy, great care must be taken to preserve the marginal artery. The mobilized pull-through segment should reach about 2 cm below the level of the pubic symphysis without tension to ensure adequate length, and resection should then be performed at least 5 cm proximal to the selected level based on rectal biopsy results to reduce the likelihood of a transition zone pull-through.

Stooling outcomes after redo pull-through surgery have not been adequately researched. However, in a previous review from Mott Children's Hospital, 32 children underwent a redo pull-through. Excluding those who were neurologically impaired, 94% had reasonable long-term stooling outcomes. [10] Outcomes in our climes may be influenced by a number of factors ranging from a paucity of surgical options and experience, delayed presentation of patients, and non-compliance to care. Finally, most patients are lost to follow-up and therefore cannot be monitored for complications.

Conclusion

This case highlights the challenge of incomplete resection in a resource-poor setting where there is no frozen section to determine the adequacy of colon resection during surgery. HD is an uncommon anomaly seen in adults and usually, evaluation and surgical correction is completed in the early years of life. However, a few populations of adults present with a residual of this condition in adulthood. This is both surgically challenging and fraught with short- and long-term morbidities. Hence, an algorithm that systematically evaluates resected bowel segments for ganglion cells must be developed for patients in resource-poor settings to prevent residual HD.

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