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Adult Hirschsprung's Disease

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Keywords: Adult Hirschsprung's disease; Refractory constipation; Swenson's pull through.

Abbreviations: HSD: Hirschsprung's Disease; CT: Computed Tomographic Scan; SAM: Severe Acute Malnutrition; MD: Doctor of Medicine; PCP: Primary Care Physician; HUCSH: Hawassa University Comprehensive Specialized Hospital.

Abstract

Background: Although the primary pathology of Hirschsprung's disease is a congenital gut motor neuron development disorder causing functional intestinal obstruction, which is diagnosed in over 94% of cases with clinical evidence and histologic examination before children reach the age of five, the term "adult Hirschsprung's disease" is conventionally used as a clinical trend when the disease is diagnosed in an adult who is older than ten years. Despite this, the disease is almost always misdiagnosed as refractory chronic functional constipation in adults.

Case presentation: In this study, we report the case of a 27-year-old female patient who was referred to a gastrointestinal referral clinic due to persistent problems of passing stool and flare-ups of her symptoms. She was receiving regular laxatives, and an enema, but there was no improvement observed on subsequent follow-ups. Consequently, adult Hirschsprung's disease was diagnosed clinically and treated with a diversion sigmoid loop colostomy with rectal and colostomy site biopsy, which confirmed HSD. The patient was treated with Swenson's pull-through technique.

Result: In the long run, the patient's long-term post-operative course was satisfactory because the biopsy-proven adult HSD was successfully managed using the same surgical principles that apply to paediatric age. Initially, diversion of the sigmoid loop colostomy resulted in a significant decrease in colonic calibre and patient improvement from secondary morbidity, which later greatly contributed to the Swenson pull through procedure and surgery-related complications.

Conclusion: Thus, in adults with a history of prolonged functional constipation, adult Hirschsprung's disease should be regarded as one of the differential diagnoses for chronic constipation, and the patient should be examined with a diagnostic histologic examination of the bowel, since Swenson's pull-through is a surgical management option that can be used as in the pediatric age group.

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Introduction

Background of study: Hirschsprung's disease is congenital absence of ganglionic cell either from failure of caudal migration of neural crest cell or failure to survive after migration of neural crest cell or failure of maturation in sub mucosal/meissner's and myenteric/Auerbach's nerve plexus result in incapable of Antegrade peristaltic propulsive movement usually presented with delayed meconium passage at birth, early neonatal obstruction, progressive abdominal distension, infrequent passage of loose stool with difficulty, significant wasting, failure to thrive in children and symptoms of chronic constipation in adults.

Statement of the problem: Lack of physician awareness in late presentation and gold standard diagnostic investigation needed for adult HSD result in disease misdiagnosis as refractory constipation which leads to delay in management and secondary morbidity.

Purpose of the study: Even though a few literature and case reports exist in other parts of the world in diagnosis and management of adult HSD there is no enough literature in Africa and Ethiopia in particular on the sequence of management in different case scenario of adult HSD and options of surgical procedure to apply with regard to its feasibility and outcome. In this case study we are describing adult HSD diagnosed at 27 years, initially diversion loop colostomy later on Swenson pull through procedure done and postoperative patient has satisfying outcome.

Objective of the study: To improve physicians' awareness on early diagnosis and surgical management options of adult Hirschsprung's disease.

Significance of the study: This worthy clinical experience will improve awareness of physician on adult HSD by considering as differential diagnosis which has a magnificent effect on early diagnosis and proper management to avoid disease related morbidity.

Scope of the study: This particular single case study states an adult HSD diagnosis and outcome of sequential management with diversion and Swenson pull through procedure.

Literature review

The first thorough description of adult Hirschsprung's disease was published by Rosin in 1950. A substantial corpus of literature has been produced since then. The precise number of incidents that were reported is unclear, as seen by the disparities in the citations of cases that have been recorded. For instance, Zhang and Ding report 300 recorded instances up until 2016, whereas Chen et al. indicate 300 reported cases up until 2006. In response, Adamou (2019) noted an increase in instances from 490 to 600 between 2009 and 2019 [1].

Despite the fact that the exact epidemiology of Hirschsprung's disease in adults is yet unknown, most reviews of clinical profiles indicate that males outnumber females 133 to 42. This is mostly because the disease is commonly disregarded in adults. The patients' ages varied from 10 to 73 years old, with a 24.1-year-old average. Although the majority of patients have been under 30 years old [2], Hirschsprung's Disease (HD) affects about one in 5000 live births, with a male to female ratio of 3:1 to 4:1 overall [3].

Most often, impaired craniocaudal migration of neuroblasts from the neural crest during the first twelve weeks of gestation is thought to be the etiology of HSD and results in functional intestinal blockage [3]. The deformity of the hindgut known as Hirschsprung's disease is caused by the absence of intramural ganglion cells in the Meissner's and myenteric plexuses, which is exhibited by the megacolon and results in functional blockage and colonic dilatation in the vicinity of the affected segment [4]. Hirschsprung's Disease (HSD) is most commonly diagnosed in childhood, with 94% of cases detected by the age of five. However, adult Hirschsprung's disease (HSD), which is defined as a diagnosis made after the age of ten, is regarded as a missed diagnosis [5]. In order to prevent related morbidity and Mortality, a high index of suspicion is required for early diagnosis [6].

A difficulty with adult HSD is that it takes longer to diagnose and treat patients, which affects morbidity and death rates. Just a small number of instances have been reported in Africa, which makes sense given the rarity of Hirschsprung's disease in adults and its challenging identification, even in the west.

Enterocolitis is not a common problem among adult patients, but the majority of them arrive with recurring constipation that can be treated by an enema or laxatives, just like in children. Due to the possibility of hypertrophied proximal innervated colons that compensate for aganglionic rectums that are distal obstructions, persons with [7] lesser condition may not receive a diagnosis until later in life. Furthermore, cathartics and enemas are common methods these individuals attempt to treat their constipation. The dilated colon eventually stops being functional in anterograde propelling of faecal matter result in retention [8].

In nearly all cases, chronic constipation is present. A biopsy is required to confirm the lack of ganglion cells, but the diagnosis may be suggested by the barium enema examination if an area of smooth narrowing with proximal dilatation is observed (83%). A dilated colon without a constricted section was present in 14% of patients [9].

To confirm the diagnosis, imaging investigations, anal manometry, and full- thickness rectal biopsies are typically carried out after Hirschsprung's disease is clinically suspected [10]. Due to the difficulty of diagnosing this illness, PCPs, gastroenterologists, surgeons, and pathologists must collaborate across specialties and have high standards for biopsies. The usual course of treatment is surgery intended to remove or bypass the aganglionic colonic or rectal segment; following surgery, quality of life can be greatly enhanced [11].

There have been reports on a number of surgical care alternatives for adult HSD in addition to diagnosis; nonetheless, there has been no specific surgical treatment or better surgical management advocated as general.

Comparison of the various operational methods for Hirschsprung's disease with regard to functional results and postoperative complications is made possible by an analysis of 199 adult instances of the disorder. The most appropriate surgical management techniques include the Duhamel, Swenson technique, the Soave endorectal pull-through procedure, and anorectal myectomy with low anterior resection [12].

Between 1950 and 1978, the Mayo Clinic performed operations on fifty adult patients with Hirschsprung's disease. There were six separate operations applied. Nonetheless, all three of the patients who underwent the Soave endorectal pull-through treatment had outstanding long-term outcomes. Two of the patients experienced anastomotic leaks and needed diverting

colostomies.

There was only one small problem for the four patients who had the Duhamel operation, and all of them had good results [13]. Due to anatomical changes, surgical therapy becomes more difficult as an adult. Several surgical techniques have been described; Swenson, Duhamel, Soave, and Lynn being the most well-known. There have been suggestions for changes to these methods. Resecting or excising the aganglionic section and lowering the healthy colon-which is ordinarily innervated at the anus-are the objectives of surgical treatment [14].

Thus, on rare occasions, Hirschsprung's at adulthood has been linked to doctors' ignorance and misinterpretation, which raises mortality and morbidity even more. This presents an anatomical obstacle for surgical care, following the same principle in the pediatric age range of under 10 years.

In the hopes that our contribution would positively influence the diagnosis and surgical treatment of adult Hirschsprung's disease, we are sharing here our clinical experience in diagnosing and treating a rare condition.

Case presentation

This is a 27year old female patient referred to our HUCSH whom had multiple health care facility visit having long standing compliant of difficulty of stool passage since she known herself as old children, with current exacerbation of progressive abdominal distension, significant weight loss but unquantified, frequent abdominal pain, appetite loss and on periodic laxative use and rectal enema, for relieving of her symptoms, objectively evaluation the patient was chronically sick looking and wasted, hard palpable faecal mass abdominally, loose stool component on digital rectal examination with rectum full faeces gaseous large bowel on, hugely distended colon on contrast enema (Figure 2) and computed tomography (Figure 1), moderate aneamia of chronic illness on complete blood count, hypokalaemia of 1.7mEq/L, hyponatremia of 120mEq/L, severe hypoalbuminemia of 1.21g/dl with all above evidence adult onset severe acute malnutrition and adult Hirschsprung's disease diagnosed clinically for which patient was managed for acute secondary complications of SAM at the same time she had advised for diversion colostomy and rectal biopsy

So that on initial intraoperative finding colon was significantly dilated having typical transition zone at recto sigmoid region (Figure 3) with multiple faecalomas in the colon seen and biopsy from rectum and colostomy site taken, diversion loop colostomy and faecaloma removal was done post operatively patient was discharged home to be on regular follow up then biopsy confirmed rectum aganglionated and colostomy site gangilonated (Figure 6 & 7)

patient significantly improved from her malnutrition associated and abdominal symptom and no colostomy related complication seen and put on waiting list for Swenson's pull through procedure.

definitive surgery was done 08 month on loop colostomy after she was counselled for procedure related complication, intraoperative due to significant weight gain intra-abdominal fat and deep female pelvis make pelvic dissection little difficult from routine pull through done in pediatric age group but we managed to do successful Swenson's pull though and the resected aganglionated bowel segment(Figure 4) specimen sent for histopathologic exam(Figure 5) and patient condition was smooth post-operative except she had superficial surgical site infection at colostomy closure site which managed with wound care and discharged and biopsy revealed same result and on 6 month follow up patient doing well having regular bowel habit.

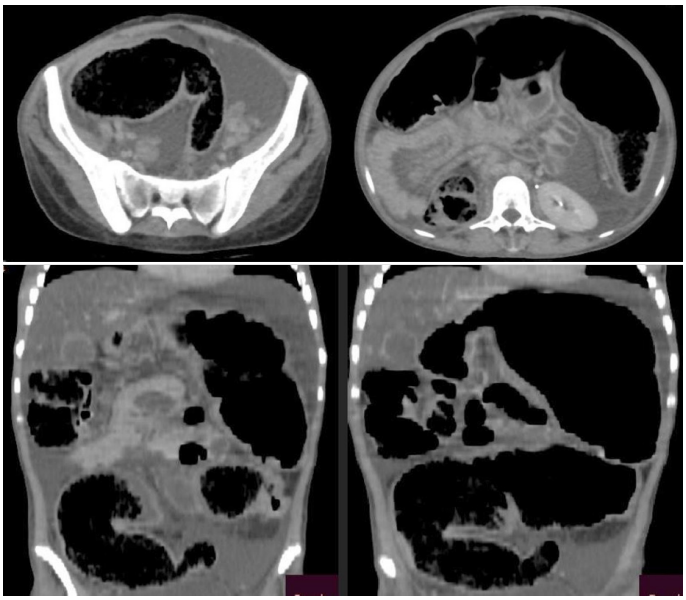

Figure 1: Axial and coronal section of abdominopelvic CT-scan with contrast shows significant colonic dilatation above transition zone and collapsed rectum below as indicated by black arrow.

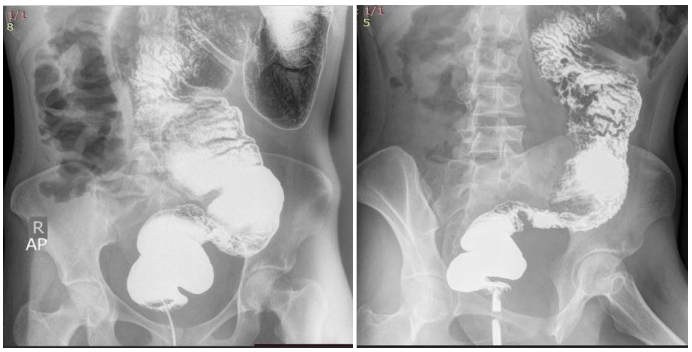

Figure 2: Contrast barium enema showing rectal collapse and significant Colon dilatation and the transition point at rectosigmoid and faecal loaded entire colon.

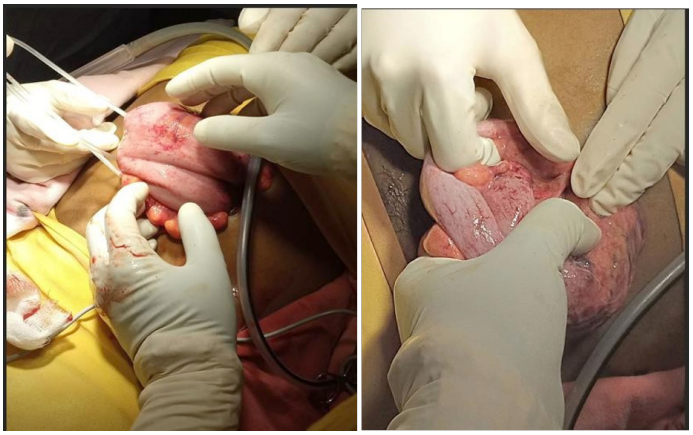

Figure 3: Intraoperative photograph shows hugly dilated sigmoid colon to delivered to wound for diversion colostomy.

Discussion

The disease's incidence is unknown, most likely due to lack of awareness lead to frequent misinterpretation, although the first reports of adult Hirschsprung's disease date back to the middle of the 1900s. In our investigation, the patient was a 27-year-old female who had no family history of a related illness.

When adult Hirschsprung's disease manifests as refractory constipation with progressive abdominal distention, a palpable hard faecal matter contrast enema can clearly show the transition zone and state the reverse ratio. However, if there is any uncertainty about other differential diagnoses, a CT scan can help rule out other conditions that could be related to malignancy. But the gold standard for diagnosing HSD is a colonoscopy that confirms the lack of ganglionic cells/hypertrophied nerve in the submucosal and myenteric enteric nervous system of gut.

Results

Even with certain anatomical challenges Adult HSD is managed according to the same principles as pediatric HSD, meaning that patients with acute obstruction, significant proximal dilatation, and secondary morbidity such as severe acute malnutrition should have protective diversion, colostomy, and rectal biopsy performed. This will allow the patient to be optimized for further definitive surgical management, which is the common goal of overall management: the resection of an aganglionated segment of bowel.

It's important to discuss our clinical experience with the patient we treated, who initially presented with significant colonic dilatation, After a diversion sigmoid loop colostomy was performed, the colon's calibre significantly decreased, and the aganglionated rectum and hypoganglionated recto sigmoid were resected after eight months and Swenson's pull through technique was done and currently our patient having normal/regular bowel habit.

Conclusion

Hirschsprung's disease should be considered a differential diagnosis in this patient who has a history of long-term chronic constipation, a faecal matter- loaded colon, and disproportionate dilatations of colonic segments that hint at barium enema study. A rectal biopsy is required to confirm this diagnosis. Swenson's pull through can be used with or without modification depending on the degree of aganglionosis and the caliber of the gangilonated colon.

Methodology

Study area: The study is carried out at the comprehensive specialized hospital of Hawassa University and the college of medicine and health sciences.

Data collection: Information gathered through direct observation, patient charts, and patient interviews.

Case analysis: For this specific case evaluation and reporting, the case analysis is based on the body of scientific literature that already exists as well as textbooks.

Ethical considerations

Informed consent: We hereby declare that the patient has provided written informed consent for the publication of her medical case for educational purposes; the patient's name has been anonymised for privacy; a copy of the consent is available

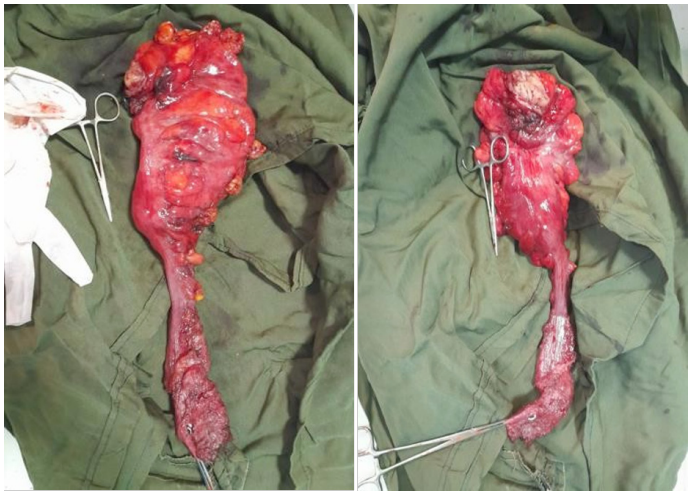

Figure 4: Intraoperative photograph of resected bowel segment shows collapsed rectum and dilated sigmoid colon with transition zone clearly seen.

Figure 5: Photograph of resected tissue specimen sent for histopathologic examination shows sample measurement.

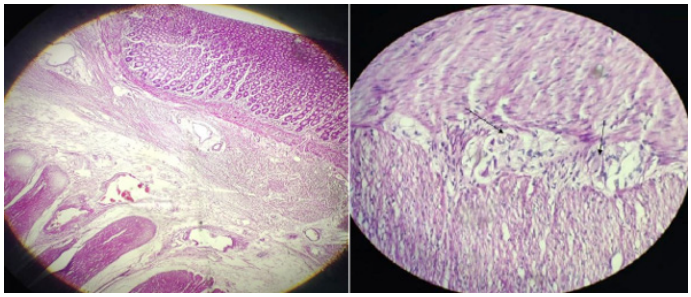

Figure 6: Low and high power section shows sigmoid colon muscularis layer having ganglion cells as shown with black arrow.

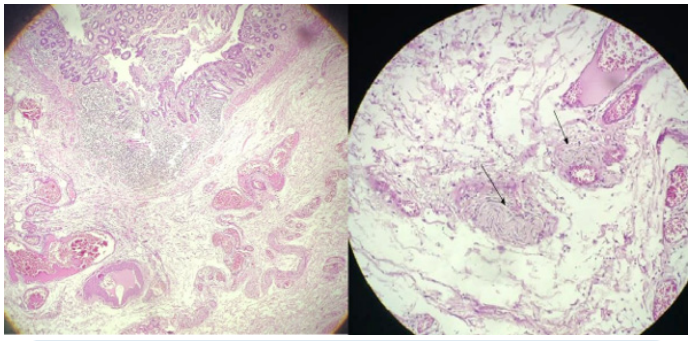

Figure 7: Low and high power-section show rectal tissue composed of bland mucosal glands and submucosa having hypertrophied nerve bundles.

for distribution to the chief editorial of this case study.

Institutional review board: We affirm that an official letter from the HUCSH institutional review board has been obtained, and it is available upon request from the main editorial of this case study.

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Disclosure of Conflict of interest: In this specific instance, we certify that there is no conflict of interest in computation.

Limitations of the study: It is challenging to describe the epidemiology and recommendations for a single instance.

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References

1. C Gamez, et al. Adult Hirschsprung's disease: A case report and literature review, Int. J. Surg. Case Rep. 2021; 82: 105881. doi: 10.1016/j.ijscr.2021.105881.

2. M Miyamoto, et al. Hirschsprung's disease in adults: report of a case and review of the literature, J. Nippon Med. Sch. 2005; 72(2): 113-120.

3. J F Qiu, Y J Shi, L Hu, L Fang, H F Wang, et al. Case Report Adult Hirschsprung's disease: Report of four cases, 2013. http://www.ijcep.com/.

4. A A Bakari, et al. Congenital aganglionic megacolon in Nigerian adults: Two case reports and review of the literature, Niger. J. Clin. Pract. 2011; 14(2): 249-252. doi: 10.4103/1119-3077.84032.

5. J Vo, R Hayler, A Tyler, K Verschuer. Chronic constipation and abdominal distension in a patient with adult Hirschprung's disease and bilateral ovarian teratomas, J. Surg. Case Reports. 2024; 4: rjae227.

6. A W Gosaye, T S Nane, T M Negussie. A case report of Hirschsprung's disease presenting as sigmoid volvulus and literature review, Tikur Anbessa Specialized Hospital, Addis Ababa, Ethiopia, BMC Surg. 2021; 21(1): 1-5. doi: 10.1186/s12893-020-00938-x.

7. A H Shitta, B T Ugwu, S D Peter, K N Ozoilo, P F Adighije, et al. Hirschsprung's disease in an adult: A case report, J. West African Coll. Surg. 2014; 4(3): 121.

8. W L F M Fayu, Chen John H, Winston III, Sanjay K Jain. Hirschsprung's disease in a young adult: Report of a case and review of the literature. 2006; 10: 347-351.

9. N L Crocker, J M Messmer. Adult Hirschsprung's disease, Clin. Radiol. 1991; 44(4): 257-259.

10. T M Rahardjo, et al. Adult Hirschsprung's disease presenting as chronic constipation: A case report, J. Med. Case Rep. 2023; 17(1): 1-5. doi: 10.1186/s13256-023-03986-y.

11. A call for awareness. A. C. R. and review of the literature Adults Hirschsprung's disease, Adults Hirschsprung's disease, a call for awareness. A Case Report and review of the literature. 2021; 79: 496-502.

12. M J Wheatley, J R Wesley, A G Coran, T Z Polley Jr. Hirschsprung's disease in adolescents and adults, Dis. colon rectum. 1990; 33(7): 622-629.

13. R A McCready, R W Beart Jr. Adult Hirschsprung's disease: Results of surgical treatment at Mayo Clinic, Dis. Colon Rectum. 1980; 23(6): 401-407.

14. H Adamou et al. Diagnosis and surgical approach of adult Hirschsprung's disease: About two observations and review of the literature. Case series, Ann. Med. Surg. 2019; 48: 59-64.

15. M I Kusuma, S Sampetoding, M Bahrun, M Faruk. Adult Hirschsprung disease as acute intestinal obstruction: A case report, Pan Afr. Med. J. 2022; 41(1).