https://dx.doi.org/10.4314/emj.v63 i14

Ethiopian Medical Journal 2025, 63(14)
Open access articles distributed in terms of
Creative Commons Attribution Licence [CC by 4.0]
ISSN 0014-1755 eISSN 2415-2420
Nigatu et al

Case report

From Obscurity to Emergence: A Rare Case of Sigmoid Colon Vovulus in Undetected Hirschsprung's Disease in Adult

Muhamad Noor Azrie Taha[1] , Siti Rahmah Hashim Isa Merican[1,2*] , Ikhwan Sani Mohamad [1,2], Maya Mazuwin Yahya[1,2], Wan Mohkzani Wan Mokhter[1,2] , Wan Hanis Wan Zakkhry[2], Wan Faiziah Wan Abdul Rahman[2,3]

[1]Department of Surgery, School of Medical Sciences, Universiti Sains Malaysia Health Campus, Kubang Kerian, Kelantan, Malaysia.
[2]Department of Pathology, School of Medical Sciences, Universiti Sains Malaysia, Health Campus, Kubang kerian, Kelantan, Malaysia
[3]Hospital Universiti Sains Malaysia, Health Campus, Kubang kerian, Kelantan, Malaysia.

*Corresponding author: rahmahkck@usm.my

Abstract
Background: *Hirschsprung Disease (HD) rarely presents in adulthood and is often overlooked due to delayed or missed diagnosis. Adult patients may present with chronic constipation, recurrent obstruction, or acute complications such as volvulus.*
Case: *We report a 20-year-old woman who presented with acute bowel obstruction and was diagnosed with sigmoid volvulus. She underwent emergency sigmoid colectomy with stoma formation and recovered uneventfully. The stoma was closed six months later, after which she remained well for nearly two years. She subsequently developed recurrent abdominal pain, distension, and constipation. Despite laparoscopic adhesiolysis for presumed adhesive obstruction, her symptoms persisted. Colonoscopy with biopsy confirmed aganglionosis consistent with very short-segment HD. Her course was complicated by Superior Mesenteric Artery syndrome, diagnosed by CT angiography, which resolved with nutritional support. She was scheduled for definitive pull-through surgery following nutritional optimization.*
Conclusion: *Adult-onset HD is rare and diagnostically challenging. Early suspicion and rectal biopsy in young adults with chronic constipation or recurrent obstruction are crucial to prevent repeated interventions and complications.*

Citation: *Taha MN, Merican SR, Mohamad IS, et al. From Obscurity to Emergence: A Rare Case of Sigmoid Colon Vovulus in Undetected Hirschsprung's Disease in Adult..Ethiop Med J 63 14*
Submission date : *14 April 2025 Accepted: 21 August 2025 Published: 30 August 2025*

Introductions

Hirschsprung Disease (HD) is a congenital disorder caused by the absence of ganglion cells in the distal gastrointestinal tract, leading to functional obstruction and proximal bowel dilatation [1,2]. It is most commonly diagnosed in infancy and represents the leading cause of neonatal intestinal obstruction [3]. In adults, HD is rare and usually reflects delayed or missed diagnosis rather than true absence of childhood symptoms. These patients often report longstanding constipation or bowel-related complaints since childhood, though the condition may only be recognized during adulthood [4].

Adult presentations are uncommon and diagnostically challenging, with chronic constipation frequently attributed to functional or idiopathic causes. This diagnostic delay is particularly common in resource-limited settings where access to specialized investigations may be restricted [3].

Sigmoid volvulus (SV) is an unusual manifestation of HD in adults, with only a small number of cases reported. A review identified approximately 31 cases in the English literature, with a prevalence of around 0.66% [5]. Among these, adult presentations are especially rare, often involving short-segment HD affecting the rectosigmoid area [6]. Misdiagnosis may result in inappropriate management such as isolated sigmoidectomy, which fails to address the underlying aganglionic segment and predisposes to recurrence or complications.

We report a case of very short-segment HD in a young woman, initially misdiagnosed as sigmoid volvulus, who underwent colectomy and later represented with recurrent symptoms. This case high-

lights the diagnostic challenges of adult HD and emphasizes the need for early suspicion and confirmatory rectal biopsy in young adults with recurrent obstruction and longstanding constipation.

Case Report

A 20-year-old woman with no prior medical history presented with a five-day history of progressive abdominal distension, colicky left iliac fossa pain, vomiting, and absolute constipation. Examination revealed a distended, tender abdomen without peritonism. Abdominal X-ray showed dilated bowel loops with a classic "coffee bean" sign, and CT confirmed a markedly dilated sigmoid colon consistent with sigmoid volvulus. Emergency laparotomy revealed a twisted, ischemic sigmoid colon with hemorrhagic intra-abdominal fluid. A sigmoid colectomy with double-barrel stoma was performed. Histopathology demonstrated ischemic changes without features of aganglionosis. Her recovery was uneventful, and the stoma was reversed six months later.

She remained well for nearly two years, after which she re-presented with recurrent abdominal distension, pain, and chronic constipation. An abdominal X-ray showed dilated bowel loops but no transition zone. She underwent laparoscopic adhesiolysis, where dense adhesions were noted, though no clear obstructive band was identified. Distal bowel loops appeared collapsed, raising suspicion of a functional obstruction.

Her symptoms persisted, with recurrent constipation, bloating, and intermittent obstructive episodes. Further evaluation with barium enema demonstrated a transition zone in the rectosigmoid region. Colonoscopy with random rectal biopsies was performed. Histopathology confirmed the absence of ganglion cells, consistent with very short-segment HD.

Following this, she developed significant postprandial pain, bilious vomiting, and weight loss. CT angiography confirmed Superior Mesenteric Artery syndrome, which was managed conservatively with total parenteral nutrition and gradual nutritional rehabilitation. Once optimized, she was scheduled for definitive anal pull-through surgery.

Discussions

Adult-diagnosed Hirschsprung Disease (HD) is exceptionally rare, with most cases recognized in infancy or childhood. Reports of adult presentations with sigmoid volvulus (SV) are even less frequent, accounting for less than 1% of HD cases [5,6]. Among the published cases, a strong male predominance has been observed, making our case in a young female particularly uncommon. Most adult patients demonstrate short-segment or very short-segment disease affecting the rectosigmoid region [6].

This patient's clinical course illustrates the diagnostic pitfalls of adult HD. Although she had a history of constipation since childhood, this was not initially recognized as pathological. Her acute presentation with sigmoid volvulus was managed with emergency colectomy and stoma formation. However, isolated resection of the sigmoid segment without addressing the underlying aganglionic bowel often leads to persistent or recurrent symptoms, as seen in this case [7].

Radiological evaluation plays an important role but is not always definitive. The "coffee bean" sign on plain abdominal X-ray is classically associated with sigmoid volvulus and is observed in 60–90% of adult cases, compared to 17–57% in children [6]. However, these figures pertain to volvulus rather than HD specifically. In suspected HD, barium enema may demonstrate a transition zone, increasing diagnostic sensitivity up to 82% in adults, though specificity remains limited [7]. Ultimately, histopathological confirmation remains the gold standard. Full-thickness rectal biopsy provides definitive evidence of aganglionosis, while immunohistochemistry with acetylcholinesterase (AChE) or calretinin staining enhances diagnostic accuracy, particularly in very short-segment disease where interpretation may be challenging [9].
Our patient's persistent symptoms following adhesiolysis raised suspicion of a functional cause, which was later confirmed histologically. Interestingly, her postoperative course was complicated by Superior Mesenteric Artery (SMA) syndrome, manifesting as postprandial pain, bilious vomiting, and weight loss, confirmed by CT angiography. SMA syndrome following HD surgery has rarely been reported but underscores the importance of careful nutritional support in this patient population.

Surgical management of adult HD requires resection of the aganglionic segment with pull-through procedures such as the Soave or Duhamel techniques [8]. Simple sigmoidectomy, as in this case, risks incomplete treatment and recurrence of obstruction. Based on our case and published reports, rectal biopsy should be considered in young patients with recurrent sigmoid volvulus or longstanding constipation to avoid missed diagnoses [7]. However, given the rarity of this condition and the limited number of published cases, such recommendations should be interpreted with caution.

This case contributes to the limited body of literature on adult HD and highlights several key lessons: the importance of thorough history-taking to elicit childhood bowel symptoms, the limitations of imaging alone in diagnosis, and the critical role of rectal biopsy in guiding definitive surgical management.

Conclusion

Adult-diagnosed Hirschsprung Disease (HD) is rare and often misdiagnosed, particularly when presenting as sigmoid volvulus. A history of chronic constipation since childhood should raise clinical suspicion in young adults presenting with recurrent obstruction. While initial surgical management may provide temporary relief, definitive diagnosis requires rectal biopsy, and treatment necessitates resection of the aganglionic segment with appropriate pull-through reconstruction.

This case highlights the importance of maintaining a high index of suspicion for HD in atypical adult presentations and illustrates the risks of incomplete treatment when the diagnosis is missed. Although the evidence base remains limited due to the rarity of reported cases, clinicians should carefully evaluate young patients with chronic constipation and sigmoid volvulus to prevent repeated interventions and improve long-term outcomes.

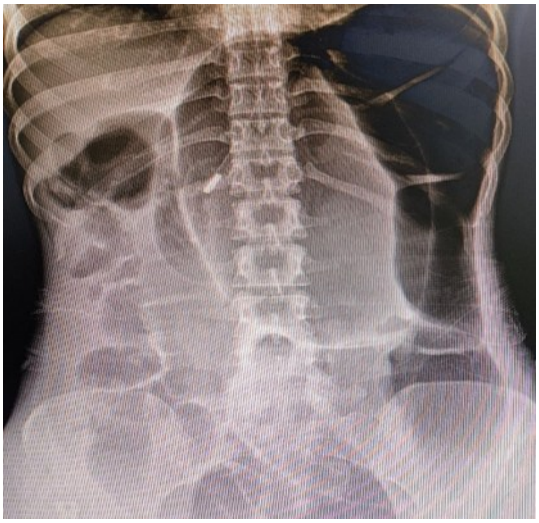

Figure 1 : Plain Abdominal x-ray, shows coffee bean signs.

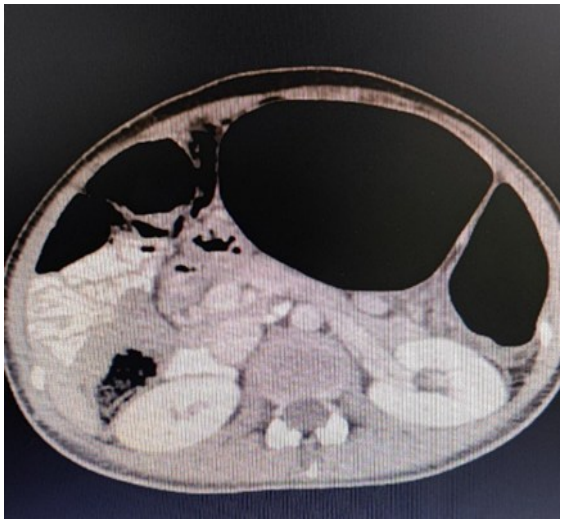

Figure 2 : Computed Tomography (CT) Scan with contrast enhanced of abdomen shows dilated sigmoid colon.

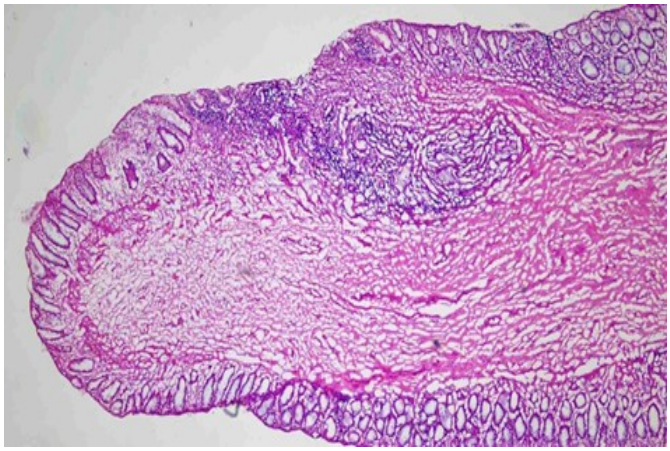

Figure 3: Frozen section of the rectal biopsy reveals absence of ganglion cells and hypertrophic nerve bundles within the submucosa layer. (4x magnification)

References

1. Chen F, Winston JH, Jain SK, Frankel WL. Hirschsprung's disease in a young adult: report of a case and review of the literature. Ann Diagn Pathol. 2006;10(6):347-351.
2. Miyamoto M, Egami K, Maeda S, Ohkawa K, Tanaka N, Uchida E, Tajiri T. Hirschsprung's Disease in Adults: Report of a Case and Review of the Literature. J Nippon Med Sch. 2005;72(2):113-120.
3. Bandré E, Kaboré RAF, Ouedraogo I, Soré O, Tapsoba T, Bambara C, Wandaogo A. Hirschsprung's disease: Management problem in a developing country. Afr J Paediatr Surg. 2010;7(3):166-168. DOI: 10.4103/0189-6725.70418.
4. Haricharan RN, Georgeson KE. Hirschsprung disease. Semin Pediatr Surg. 2008;17(4):266-275.
5. Zain M, Abouheba M. Sigmoid volvulus; a rare complicated presentation of Hirschsprung's disease: A case report. Int J Surg Case Rep. 2021;89.
6. Zeng M, Amodio J, Schwarz S, Garrow E, Xu J, Rabinowitz SS. Hirschsprung disease presenting as sigmoid volvulus: a case report and review of the literature. J Pediatr Surg. 2013;48(1):243-246.
7. Ghaemi M, Bahar MM, Motie MR, Hiradfar M, Soltani E, Saremi E. Late presentation of Hirschsprung's disease as sigmoid colon volvulus: report of four cases and review of the literature. Colorectal Dis. 2010;12(7):704.Top of Form
8. Shitta AH, Ugwu BT, Peter SD, Ozoilo KN, Adighije PF, Omolabake BI. Hirschprung’s Disease In An Adult : A Case Report. J West Afr Coll Surg. 2014;4(3):121-126.
9. Kapur RP. Calretinin-immunoreactive mucosal innervation in very short-segment Hirschsprung disease: a potentially misleading observation. *Pediatr Dev Pathol*. 2014;17(1):28-35. doi:10.2350/13-10-1387-OA.1