



INTISARI SAINS MEDIS

Published by Intisari Sains Medis

Constipation that needs attention: late Hirschsprung disease



CrossMark

Kristin Agustina^{1*}, Ni Nyoman Margiani², Pande Putu Yuli Anandasari², Ni Made Mahastuti³¹Radiology Resident, Faculty of Medicine, Universitas Udayana-Sanglah General Hospital, Bali-Indonesia²Radiology Department, Faculty of Medicine, Universitas Udayana-Sanglah General Hospital, Bali-Indonesia³Pathological Anatomy Department, Faculty of Medicine, Universitas Udayana-Sanglah General Hospital, Bali-Indonesia

*Corresponding author:

Kristin Agustina;
Radiology Resident, Faculty of Medicine, Universitas Udayana-Sanglah General Hospital, Bali-Indonesia;
yohana@gmail.com

Received: 2020-10-24

Accepted: 2021-03-15

Published: 2021-04-01

ABSTRACT

Background: Hirschsprung disease (HD) or congenital aganglionic megacolon is an intestinal motor disorder that occurs in approximately 1 in 5000 live births. It makes colon fail to relax, mainly producing the symptoms of constipation. Approximately 90% of cases are diagnosed in the first year of life. Most of the remaining 10% are made in early childhood, with less than 1% being made in teenagers or adulthood.

Case report: A 13 years old girl reported to the pediatric surgical outpatient department complaining constipation since birth. There was no history of delayed passage of meconium. A contrast enema study with water-soluble contrast showed high probability of

ultrashort segment HD. Histopathological examination revealed neither nerve fibers with ganglion cells nor hypertrophy of nerve fibers were seen confirming the aganglionosis. Surgical treatment was performed with good clinical progression.

Conclusion: Despite of its rarity, the possibility of HD should be considered in teenagers with chronic refractory constipation, especially when there was a history of delayed or non-passage of meconium after birth. An accurate diagnosis is mainly based on collective assessment of medical history, clinical examination, contrast enema study, and rectal biopsy as a gold standard.

Keywords: Hirschsprung disease, megacolon, congenital, constipation.

Cite This Article: Agustina, K., Margiani, N.N., Anandasari, P.P.Y., Mahastuti, N.M. 2021. Constipation that needs attention: late Hirschsprung disease. *Intisari Sains Medis* 12(1): 64-67. DOI: 10.15562/ism.v12i1.845

INTRODUCTION

Hirschsprung Disease (HD) is an intestinal motor disorder caused by an absence of ganglion cells in the submucosal (Meissner) and myenteric (Auerbach) neural plexuses in a variable bowel segment. It makes colon fail to relax, mainly producing the symptoms of constipation.¹⁻³

HD occurs in approximately 1 in 5000 live births with an overall male to female ratio of 3:1 to 4:1 and accounts for a substantial proportion of cases of neonatal bowel obstructions.²⁻⁶

Approximately 90% of cases are diagnosed in the first year of life, mostly during the neonatal period. Most of the remaining 10% are made in early childhood, with less than 1% being made in older children and event adult patients.^{1,3,4,7} This case is being reported because of its relative rarity.

CASE REPORT

A 13 years old girl, coming from rural area in Bali, reported to the pediatric surgical outpatient department complaining constipation and abdominal distension since birth. Her mother said that her daughter's defecation frequency is once a month. The symptoms were relieved by occasional use of laxatives and enema. There was no history of delayed passage of meconium, hematochezia and any previous surgery. The family history was unremarkable.

Physical examination revealed no abdominal mass on palpation but the abdomen was distended with fullness at the flanks. The abdomen returns a tympanic note on percussion. Rectal examination revealed good sphincter tone with a high rectal fecal load.

Routine laboratory evaluation, including a complete blood count, liver and renal function test, urinalysis, and blood chemistry studies, were within



Figure 1. Abdominal plain radiograph (BOF)

normal limits. Plain abdominal radiograph (BOF) revealed grossly dilated large bowel on right abdomen and pelvic cavity with extensive fecal loading, suspect megacolon

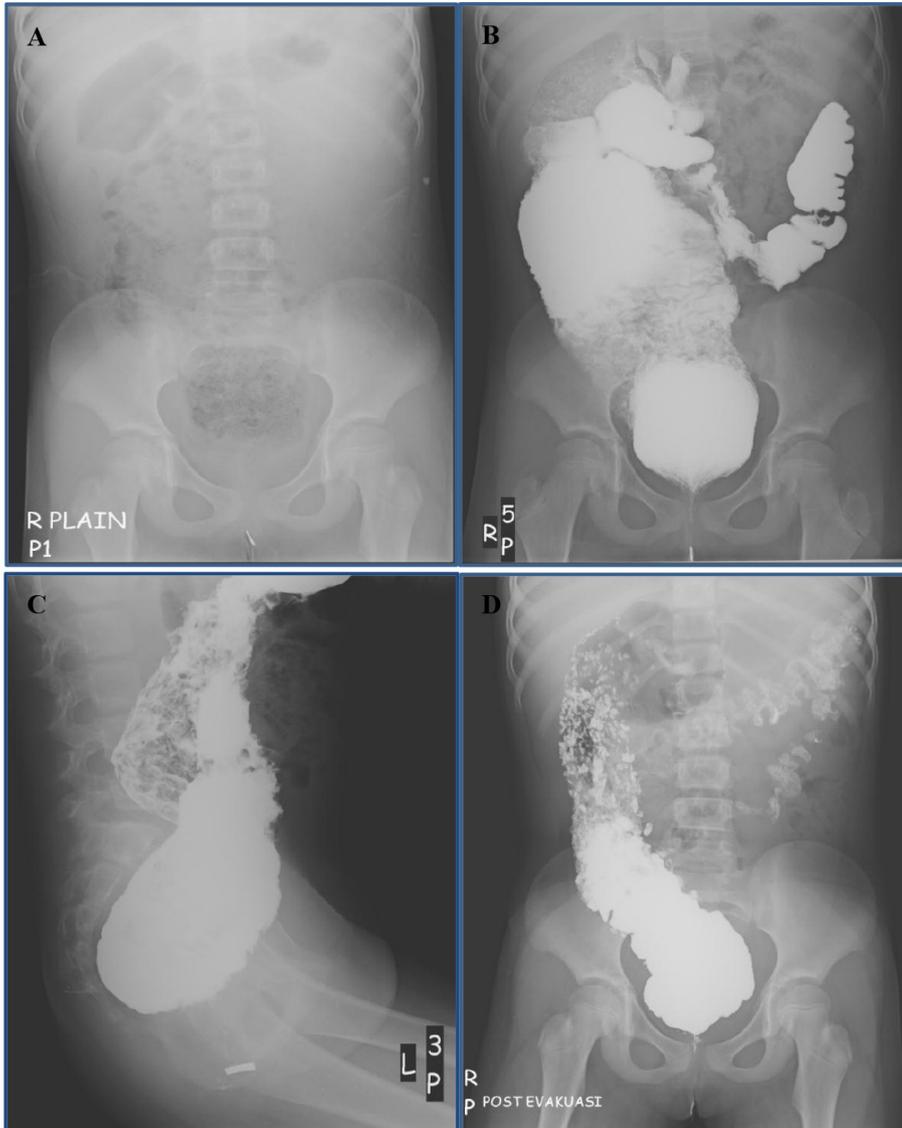


Figure 2. A contrast enema study with water-soluble contrast: **A.** Plain AP, **B.** Contrast AP, **C.** Contrast Lateral, **D.** Post Evacuation.

with fecal material retention (Figure 1).

A contrast enema study with water-soluble contrast showed: mixture of contrast with fecal matter in a dilated colon (mottled sign), rectosigmoid index (RSI) < 1 with irregular rectosigmoid mucosa, abrupt type transitional zone, no irregular contractions, the most distal contrast distance with a marker approximately 3.7 cm, and there was contrast retention with antiperistaltic on post evacuation study. These findings were considered consistent with high probability of ultra-short segment HD (Figure 2A-D).

Histopathological examination of full-thickness rectal biopsy taking about 1.5-2 cm proximal the dentate line (the

distal rectum does typically not have ganglion cells) revealed neither nerve fibers with ganglion cells nor hypertrophy of nerve fibers were seen confirming the aganglionosis (Figure 3).

Surgical treatment was performed with laparoscopic-assisted transanal endorectal pull-through (LATEP) procedure and laparoscopic assisted anorectoplasty (LAARP). The surgery consisted of releasing megacolon from mesocolon, with anoplasty and pull-through of megacolon. Megacolon resected approximately 10 cm from anal. The patient was discharged 20 days after operation and was followed up for 2 years with good clinical progression. The defecation was satisfactory (once

a day without laxatives) and without complications.

DISCUSSION

Hirschprung disease (HD) or congenital aganglionic megacolon is an intestinal motor disorder, which is caused by the failure of neuroblasts originating from the neural crest (precursors of enteric ganglion cells) to migrate completely during intestinal development in the first 12 weeks of gestation.^{1-4,7} As a result, it shows an absence of ganglion cells in the submucosal and myenteric neural plexuses in a variable bowel segment, principally affecting the rectosigmoid or rectal segment, causing a functional obstruction and mainly producing the symptoms of constipation. The other symptoms that lead to an early suspicion of HD are defined in the newborn as a greater than 48 hours delay in the elimination of meconium associated with abdominal distension and vomiting.^{3,5-7} Abdominal plain radiographs usually show large bowel obstruction image with massive distention of the proximal region of the large bowel filled with fecal material, consistent with distal intestinal obstruction.^{2,3}

In a majority of cases, HD is characteristically manifested at birth and diagnosis is made in infancy and early childhood. HD in teenager or adult patient is uncommon and often undiagnosed or misdiagnosed, as our patient who was diagnosed at 13 years old.^{1,3,4,7} Our patient had a lifelong history of constipation because the proximal innervated colon can be hypertrophied, eventual dilated (megacolon), to compensate for the distal obstructed aganglionic colon or rectum, as was demonstrated in this patient's contrast enema radiograph.^{1,7} In addition, she lives in the rural area far from medical facilities. They often try to relieve the constipation by using laxatives and local herbs. These factors contributed to delay in her diagnosis, made the doctor out of a HD hypothesis.

It is more difficult to diagnose HD in adult than in early infant. This is because HD is rare in adults, there is a higher incidence of short or ultra-short aganglionic segment and in the early stages of the disease there are relatively mild symptoms. Adult constipation and

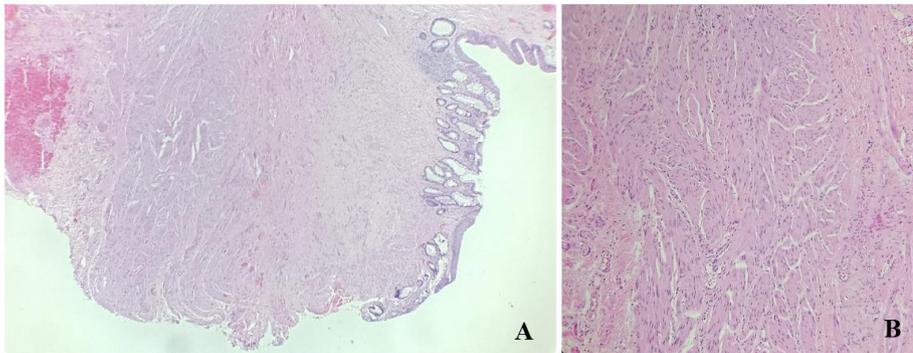


Figure 3. A. Pieces of tissue walls of the intestine consists of mucosal, submucosal and muscular layers. The mucosal layer appears to be coated in a columnar epithelium with cell goblets that are connected with squamous metaplasia epithelium. No visible picture of nerve fibers with ganglion cells or hypertrophy of nerve fibers. (H&E staining 40X) B. Muscularis layer, no visible ganglion cells. (H&E staining 100x).

acquired megacolon may be caused by neoplasm, volvulus, stricture, slow colonic motility, Chagas disease, anatomical or functional outlet obstruction, or idiopathic (non-Hirschsprung) megacolon. Other causes are dietary factors, medications, psychological factors and systemic diseases.¹ An accurate diagnosis of HD in teenagers is based on the collective assessment of medical history, contrast enema test results, and full-thickness rectal biopsy findings as the golden standard diagnosis of HD.^{3,5} Anal manometry that demonstrates the absence of internal anal sphincter relaxation upon rectal distention is not primarily used in Indonesia for diagnosis of HD.⁸

Contrast enema performed without a rectal balloon demonstrates typical finding of a transition zone between the distal aganglionic segment that is narrowed and the proximal colon that is dilated with normal ganglion cells, irregular colonic contractions, irregular mucosa suggesting enterocolitis, and an abnormal rectosigmoid index (RSI).^{1,6} Other reliable sign in contrast enema is contrast retention mixed with stool in the rectum and/or large bowel for >24 hours following a study, which clearly demonstrated in this patient's radiograph.^{1,3} In about 20% of the patients with adult HD, a dilated colon without characteristic rectal narrowing, as seen in our patient, is demonstrated. This finding may be due to a short or, more commonly, an ultrashort diseased segment.¹ Do not take a digital rectal

maneuvers for examination or irrigation before the contrast enema because it can lead to a false-negative radiologic result.³ The result of the histopathological evaluation is characterized by the absence of ganglion cells in the submucosal and myenteric neural plexuses and also the presence of hypertrophied nerve trunks in the space usually occupied by the ganglion cells.^{3,5,6}

Computed Tomography (CT) is more useful for better anatomic portrayals, view the dilated colon and the transition zones, and to definitively exclude other diseases which can also cause chronic constipation, such as colorectal cancer. But there is a risk of excessive radiation and more expensive methods.^{1,5} Our patient had no need for CT scan as the contrast enema was diagnostic.

According to the length of aganglionic colon, HD is classified into 4 categories. Short aganglionic segment (75-80% of cases), when the aganglionic segment is present in the distal part of the sigmoid colon and rectum. Long aganglionic segment (10%), when it outruns up to the splenic flexure. The rarest form of HD with the most severe clinical course is total colonic aganglionosis (5%) that affect the entire colon. The last form is ultra-short aganglionic segment that involves only the distal part of the rectum, anal canal above the pectinate line.⁹ Adolescents and adults who suffer from intractable chronic constipation may have short segment or ultra short segment Hirschsprung's

disease. (Amel A. Hashish). The case reported here was ultra short segment HD.

After HD is diagnosed, the management is usually surgery.⁵ The principle surgical management of HD is to remove the aganglionic segment and pull-through the proximal normally innervated bowel. This definitive treatment first described by Swenson and Bill in 1948.¹⁰ Corrective surgical techniques have evolved over time. Duhamel and Soave described the retro-rectal pull-through and endorectal pull-through respectively. The outcome and prognosis of these three surgical techniques (Swenson, Duhamel, and Soave) have been very good in the improvement of patients.⁷

CONCLUSION

Despite of its rarity, the possibility of HD should be considered in teenagers with chronic refractory constipation, especially when there was a history of delayed or non passage of meconium after birth. An accurate diagnosis is mainly based on collective assessment of medical history, clinical examination, contrast enema study, and rectal biopsy as a gold standard.

FUNDING

The author states that he gets funding independently without involving any sponsors.

AUTHOR CONTRIBUTION

All authors have the same contribution in the preparation of this research report, from the preparation of a conceptual framework, data collection, analysis of research data, to the preparation of a publication manuscript in the form of a research report.

CONFLICT OF INTEREST

The Authors declare there is no conflict of interest regarding publication of the case report.

PUBLICATION ETHICS

The patients or parents had received written signed informed consent regarding publication of medical data in medical scientific journal.

REFERENCES

1. Suleiman Lawal, Philip O Ibinaiye, Sefiya A Olarinoye-Akorede, Joseph B Igashi AHU. Case Report Late diagnosis of Hirschsprung's disease. Published online 2017:70-73. doi:10.4103/ssajm.ssajm
2. Qiu J, Shi Y, Hu L, Fang L, Wang H, Zhang M. Case Report Adult Hirschsprung's disease : report of four cases. 2013;6(8):1624-1630.
3. Prabhakaran S. Hirschsprung's disease. Int J Acad Med. 2017;3(3):96-100. doi:10.4103/IJAM.IJAM_12_17
4. Bakari AA, Gali BM, Ibrahim AG, 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. Estopiñan Rebolgar R, Estopiñan Cánovas R, Pila Peláez R. Hirschsprung's disease in an adult patient. Rev Colomb Gastroenterol. 2016;31(1):52-55. doi:10.22516/25007440.73
6. Alehossein M, Roohi A, Pourgholami M, Mollaeian M, Salamati P. Diagnostic Accuracy of Radiologic Scoring System for Evaluation of Suspicious Hirschsprung Disease in Children. Iran J Radiol. 2015;12(2):0-4. doi:10.5812/iranradiol.12451
7. Garg SR, Sathe PA, Taware AC, Surve KM. Fatal toxic megacolon in a child of Hirschsprung disease. J Clin Diagnostic Res. 2016;10(12):ED03-ED05. doi:10.7860/JCDR/2016/21075.9083
8. Indonesian Ministry of Health. National Guide on Hirschsprung. Kepmenkes HK0107/MENKES/474/2017. 2017;6:5-9. [Cited March 02nd 2020]. Available at: <http://hukor.kemkes.go.id/hukor/kepmenkes/7/221>
9. Szyllberg L, Marszałek A. Diagnosis of Hirschsprung's disease with particular emphasis on histopathology. A systematic review of current literature. Prz Gastroenterol. 2014;9(5):264-269. doi:10.5114/pg.2014.46160
10. Swenson O, Bill AHJ. Resection of rectum and rectosigmoid with preservation of the sphincter for benign spastic lesions producing megacolon; an experimental study. Surgery. 1948;24(2):212-220.



This work is licensed under a Creative Commons Attribution