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Malnutrition and Developmental Delay Presenting as Complications of Late Onset Hirschsprung's disease : A Case Report

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ABSTRACT

Background: Hirschsprung's Disease is a congenital disorder characterized by the absence of ganglionic cells in the colon, causing loss of peristaltic movement in the bowel. Most cases are diagnosed before the age of one and marked by the delayed passage of meconium, vomiting, and distended abdomen. Here, we present a case of a 2-year-old boy with an atypical presentation of malnutrition and developmental delay as a complication of Hirschsprung's Disease..

Case Presentation: A 2-year-old boy who presented with abdominal distention and chronic constipation was referred to our hospital. Past personal history revealed that he frequently suffered from the symptoms but was previously diagnosed with regular constipation and only consumed symptomatic medication. On admission, his vital signs were all normal, but his appearance was pale and weak. Growth and developmental screening showed poor growth status and developmental delay in all four domains. From the physical examination, gross abdominal distention was found and the rectal examination demonstrated a tight anal sphincter, empty rectal vault, and a green-colored stool with no forceful expulsion. Radiologic and histopathological findings suggested Hirschsprung's disease.

Conclusion: Chronic abdominal distention followed by malnutrition and developmental delay can present as a complication of hirschsprung's disease in older children. Due to these subtle signs and symptoms, a thorough examination and proper diagnostic algorithm are essential to assess patients' clinical condition without missing the possibility of rare congenital disease.

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1. Introduction

Hirschsprung's disease is a congenital disorder with an incidence of 1 to 2 cases per 10.000 live births. It typically presents in the newborn baby, but in some cases, it is found in older children and adults with atypical presentation. Many factors contribute to the delay of the diagnosis, but the diagnosis of Hirschsprung's Disease in older children and adults is mainly caused by a lack of awareness of the symptoms and difficulties in accessing quality care. A high index

of suspicion is necessary to investigate the problems and prevent morbidity and mortality.

In this study, we report a case of a 2-year-old boy with abdominal distention and was found to have severe malnutrition and developmental delay due to his condition.

2. Case presentation

A 2-year-old boy was referred to Sultan Agung Islamic Hospital with symptoms of abdominal distention and chronic constipation. His past personal



Figure 2. A 2-year-old boy with gross abdominal distention.

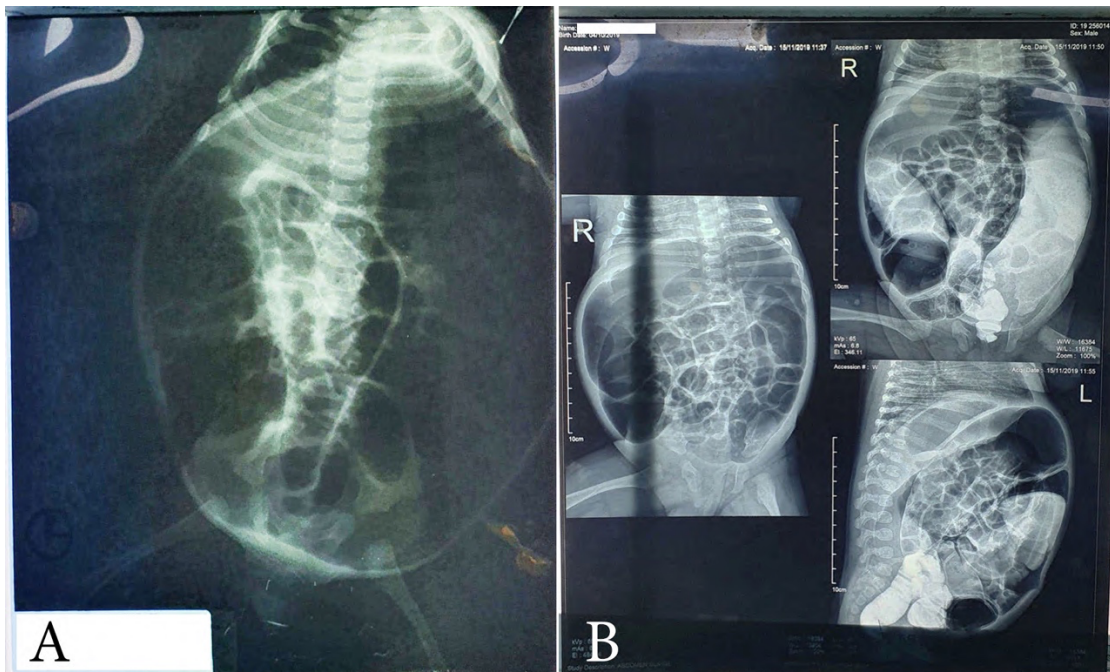


Figure 3. A. Dilated small intestine and colon filled by massive fecal material on plain radiograph. There was no free air seen. B. Colon in loop examination demonstrated a reduced caliber of the distal rectum and sigmoid colon (1,55cm in length approximately) with dilation of its proximal part.

3. Discussion

This study reports a late-onset of Hirschsprung's Disease with malnutrition and developmental delay as its complication. Hirschsprung's disease is commonly considered a newborn disease, but certain circumstances in developing countries may lead to a late diagnosis or inadequate medical treatment¹. Ignorance of the patient, lack of knowledge, and

habitual long-term of chronic constipation usually become the main reason for the delayed diagnosis². Delayed passage of meconium, vomiting, and distended abdomen is most commonly found in infants and been associated with higher mortality rate³. Early diagnosis is crucial in preventing later complications. Late-onset hirschsprung's disease (in children of 2 years age or over) usually comes with a different presentation than in the newborn period^{2,4}.

Symptoms in older children are more subtle, it usually comes with various and more complex complications, like Hirschsprung-associated enterocolitis, chronic constipation, failure to thrive, or more⁵. However, short and long-term complications remain a considerably serious problem. In this case, the patient came with abdominal distention and chronic constipation followed by malnutrition and developmental delay. In this study, the patient suffered from chronic obstruction, sometimes he was unable to defecate at all so the patient's mother had to place a rubber tube to help him defecate, but some other times the stool was able to pass a bit.

The patient's poor feeding is responsible for his malnutrition and developmental delay. Malnutrition can be a serious problem. Chronic obstruction affects the patient's digestion process and nutrient absorption, including iron and other materials needed for blood formation. As a result, older children with hirschsprung's disease often experience a nutritional deficiency, growth retardation, and anemia⁶. In this study, the patient had a severe anthropometric profile, where all of his Z-Scores were -4 SD or below (normal range: +2 until -2 SD).

Khandelwal et.al revealed that a Z-Score of less than three SD and the presence of anemia were significantly associated with developmental delay⁷. In the same way, other previous studies revealed that under-nutritional status, that is being underweight, stunting, and wasting, among under-five children was significantly associated with developmental delay⁸. These studies are in line with our findings that the patient underwent a delay in four major domains of development: gross-motor, fine-motor, language, and personal-social domain. Malnutrition is a potential risk factor for neurodevelopment. Brain development is sensitive to nutritional deficits. Different nutrients preferentially affect different brain functions⁹.

These problems can be the presenting symptom in some hirschsprung's disease patients, and may not be immediately recognized due to the rarity of the disease. Delay in diagnosis remains a challenge in developing countries because it may potentially affect clinical outcomes. Late referral and inadequate access to specialized health facilities are the main reasons for late diagnosis. Moreover, only 90% of all hirschsprung's diseases present typical clinical features during the newborn period¹⁰. Hence, a thorough physical examination and comprehensive look at the patient's past medical illness should be conducted to open up the possibility of hirschsprung's disease as soon as possible.

Moreover, malnutrition and anemia were known as potential risk factors for anastomotic leakage

after surgery. A study conducted by Ouladsaiad revealed that operative strategies in older children were more difficult, especially in malnutrition conditions. Fortunately, we succeeded to perform the colostomy safely with no difficulties¹⁰. However, the patient was advised to do a routine follow-up to make sure he has no significant surgical complications and his nutritional status is improved.

4. Conclusion

Chronic abdominal distention followed by malnutrition and developmental delay can present as a complication of hirschsprung's disease in older children. Clinicians should assess their patient's clinical presentation & past illness comprehensively to open up the possibility of hirschsprung's disease and plan the best treatment

Conflicts of interest

The authors declare no conflict of interest

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Authors' contributions

PA conceptualized the first draft. RY performed the operation and critically revised the technique. SZO wrote the manuscript and provided the data needed. All authors read and approved the final manuscript.

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Ethical Approval

This case report has obtained ethical approval from the Health Research Ethics Committee, Faculty of Medicine, Sultan Agung Islamic University, Semarang with the number 149/IV/2023/Komisi Bioetik

Reference

1. Mabula JB, Kayange NM, Manyama M, Chandika AB, Rambau PF, Chalya PL. Hirschsprung's disease in children: a five year experience at a University teaching hospital in northwestern Tanzania. *BMC Res Notes*. 2014 Dec;7(1):410.
2. Adamou H, Amadou Magagi I, Habou O, Adakal O, Aboulaye MB, Robnodji A, et al. Diagnosis and surgical approach of adult Hirschsprung's disease: About two observations and review of the literature. Case series. *Annals of Medicine and Surgery*. 2019 Dec;48:59–64.

3. Granéli C, Dahlin E, Börjesson A, Arnbjörnsson E, Stenström P. Diagnosis, Symptoms, and Outcomes of Hirschsprung's Disease from the Perspective of Gender. *Surgery Research and Practice*. 2017;2017:1–8.
4. Soussan H, Jabi R, Ouryemchi M, Haddadi Z, Bouziane M. Hirschsprung's Disease in Adults Revealed by an Occlusive Syndrome. *Cureus* [Internet]. 2021 Oct 4 [cited 2023 Mar 15]; Available from: <https://www.cureus.com/articles/71057-hirschsprungs-disease-in-adults-revealed-by-an-occlusive-syndrome>
5. Apte A, McKenna E, Levitt MA. Image of the Month: Decision-Making in Surgery for Late Onset Hirschsprung Disease. *European J Pediatr Surg Rep*. 2020 Jan;08(01):e99–101.
6. Sun X, Chu J, Li C, Deng Z. Hirschsprung's disease presenting as intractable anemia: a report of two cases and review of the literature. *BMC Pediatr*. 2020 Dec;20(1):525.
7. Khandelwal N, Mandliya J, Nigam K, Patil V, Mathur A, Pathak A. Determinants of motor, language, cognitive, and global developmental delay in children with complicated severe acute malnutrition at the time of discharge: An observational study from Central India. Lazzerini M, editor. *PLoS ONE*. 2020 Jun 1;15(6):e0233949.
8. Wondemagegn AT, Mulu A. Effects of Nutritional Status on Neurodevelopment of Children Aged Under Five Years in East Gojjam, Northwest Ethiopia, 2021: A Community-Based Study. *IJGM*. 2022 Jun;Volume 15:5533–45.
9. Chattopadhyay N, Saumitra M. Developmental Outcome in Children with Malnutrition. *J Nepal Paediatr Soc*. 2016 Dec 31;36(2):170–7.
10. Ouladsaiad M. How to manage a late diagnosed Hirschsprung's disease. *Afr J Paediatr Surg*. 2016;13(2):82.