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Case Report

Ultrashort segment adult Hirschsprung disease: A case report of periodic abdominal distension and constipation spanning for more than 20 years ☆

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ABSTRACT

The diagnosis of adult Hirschsprung disease (HD) is rare, furthermore there are only 3 or 4 cases of adult ultrashort segment HD (USHD) reported previously in English literature to our knowledge. Herein, we present a case of a 22-year-old female presented with long standing history of constipation and abdominal distension secondary to USHD. Imaging modalities included plain abdominal X-ray, ultrasound (USG) and computed tomography (CT) scan which collectively aided in establishing the diagnosis. Through this case report, we aim to emphasize that HD should be suspected in every adult population with chronic constipation. Also, it highlights the role of CT scan as how it should be the preferred diagnostic tool for suspected adult HD, as, not only it helps correctly identify the transition zone but rule out other possible causes of chronic obstruction.

Introduction

Hirschsprung disease (HD) is characterized by absence of ganglion cells in distal bowel commonly seen in infants and children who present with features of obstruction like abdominal distension, constipation and occasional bilious vomiting. The differential diagnoses for large bowel obstruction in children and neonates are limited, but in adults, the list expands significantly. Most of the time in our clinical setting, HD is not even suspected owing to its rarity in adult population. Moreover, occasional defecation in ultrashort-segment Hirschsprung disease (USHD) makes the diagnosis more challenging [1]. Detailed patient history, appropriate radiological investigations and histopathological findings are necessary for correct diagnosis of this condition. We present a case of ultra-short segment adult HD and describe the various imaging features in this case report.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Case presentation

A 22-year-old female complained of unable to pass feces and flatus for the past 2 days with multiple (about 20-25) episodes of vomiting. This was followed by progressive abdominal distension and pain. Past history revealed similar such episodes of chronic constipation that afflicted her since birth which was managed by laxatives and manual de-impaction. Her last visit for similar symptoms to the hospital was 9 years ago where she was managed conservatively, however she has been taking oral laxatives for constipation on a regular basis. On, physical examination patient was afebrile but ill-looking, was hemodynamically stable with a large distended abdomen. Clinical examination revealed a distended, firm, nontender abdomen without local rise in temperature with tympanic note on percussion. The case was shifted to the ICU with provisional diagnosis of large bowel obstruction. Abdominal radiographs were requested which showed a markedly dilated colon occupying abdominal cavity showing mottled appearance (Fig. 1). Bedside ultrasound (USG) was performed, revealing a distended rectum. However, it was of limited utility as air from the distended bowel obscured visualization of other structures. Upon administration of enema, symptoms were alleviated and distension was reduced. CT scan was performed, showing a massive dilatation of sigmoid colon (largest diameter of 23 cm) and rectum with internal mottled gas pattern and a large fecaloma (Figs. 2 and 3). Dilated sigmoid colon was seen compressing and displacing liver to the right upper quadrant, spleen and stomach into the left upper quadrant. Smooth funnel-like tapering was noted towards the anus approx 2.4 cm from anal verge corresponding to transitional zone. Peri-rectal fat planes appeared normal. No obvious intraluminal bowel pathology, wall thickening or abnormal contrast enhancement was noted. A full-thickness biopsy of rectum was obtained 2 cm proximal to the dentate line. Tissue examination revealed a ganglionosis, establishing a diagnosis of ultra-short segment Hirschsprung disease (Fig. 4).

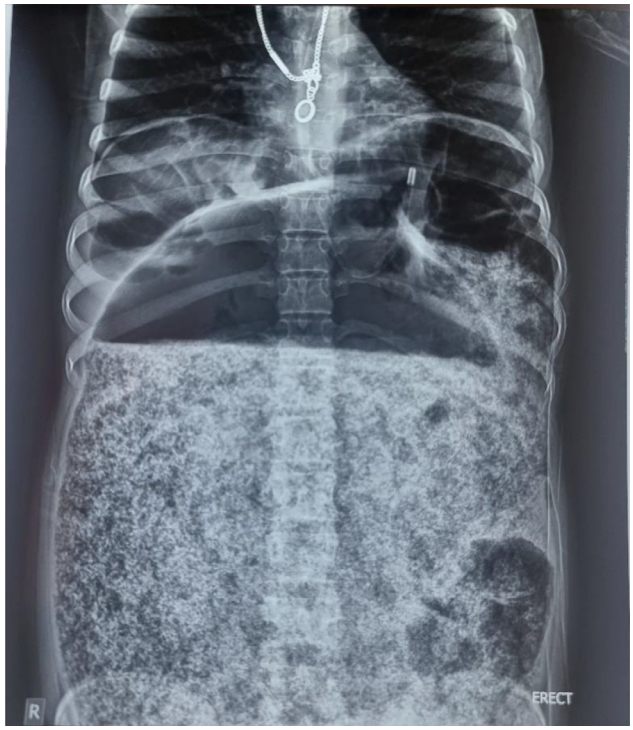

Fig. 1 – Abdominal X-ray shows markedly dilated colon occupying the abdominal cavity with mottled gas appearance corresponding to fecaloma.

Discussion

Although relatively common in young age group, HD in adults is often a rare diagnosis. Clinicians, initially consider malignancy, diverticulitis or volvulus as potential causes when examining a case of abdominal distension and constipation.

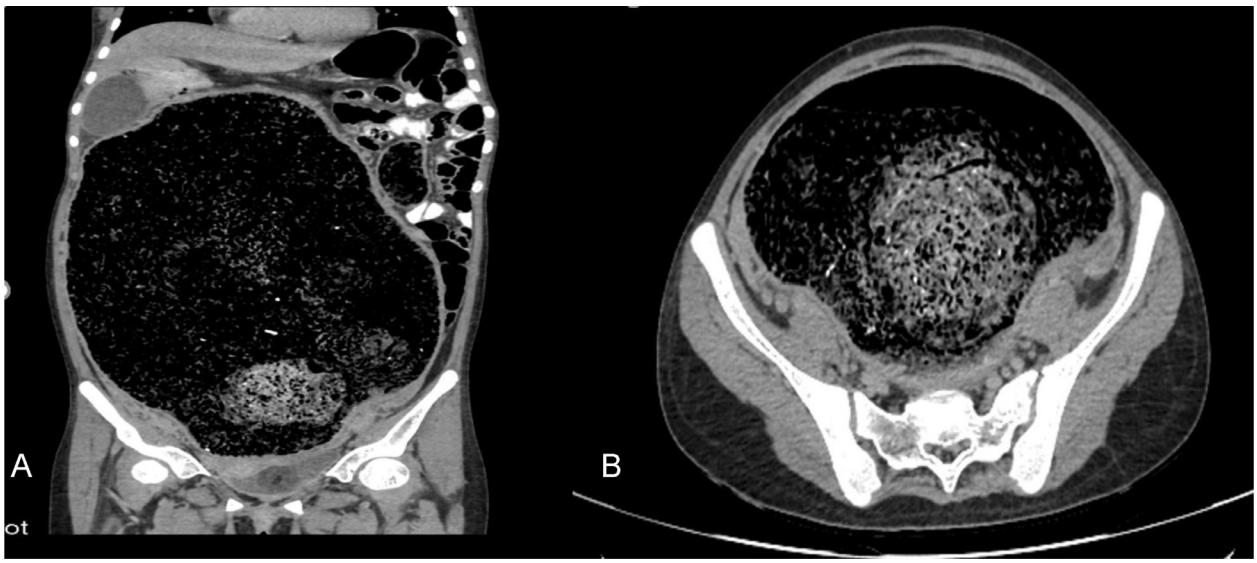

Fig. 2 – Coronal (A) and axial CT scan (B) section shows grossly dilated sigmoid colon with fecal component within. Also, the dilated colon is seen compressing and displacing surrounding structures.

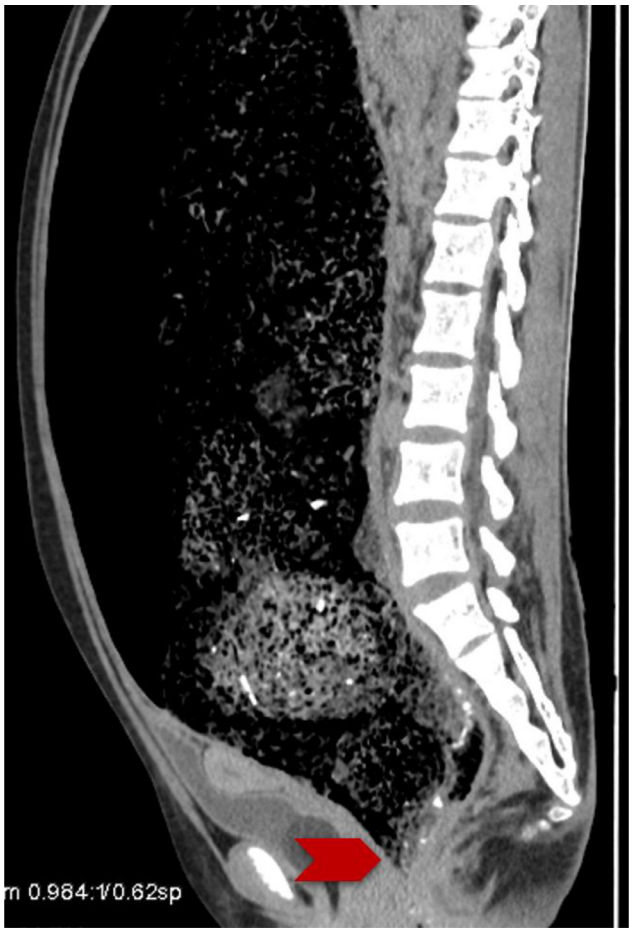

Fig. 3 – MDCT in sagittal reformatted image shows megarectum and fecaloma along with the typical funnel shaped transition zone (red arrowhead). TZ was approx 2.4 cm from anal verge.

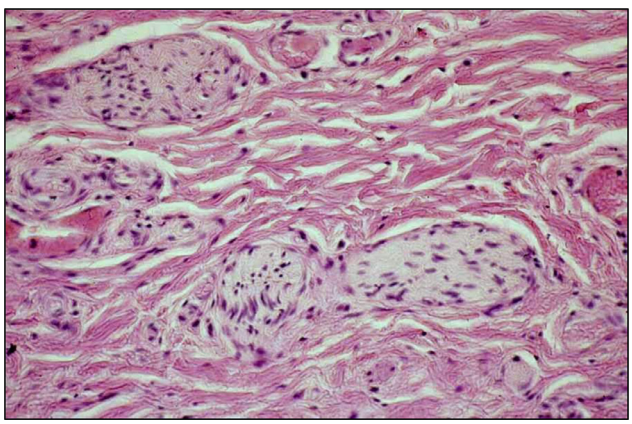

Fig. 4 – Hypertrophied nerve bundle with collagenization and devoid of any ganglion cells.

In young adults, especially in Africa and Asia, the most common cause of large bowel obstruction is volvulus [2]. There is a diagnostic challenge surrounding HD. Firstly, the diagnosis of HD is often missed in adults. Secondly, occasional relief of symptoms in USHD variant complicates diagnosis. And, thirdly, once symptoms are alleviated with laxatives, patients may resist undergoing invasive procedures. These 3 factors collectively influence subsequent patient management.

Chronic constipation that is obstinate to treatment, abdominal distension and pain with a palpable abdominal mass are commonly encountered symptoms in adult HD. Despite HD commonly affecting male patients with a ratio of 4:1 [1], in our instance, it presented in a female patient.

Barium enema is commonly used in both children and adults for diagnosing HD. However, due to its contraindications, limited ability to rule out other conditions, and the potential for false positive results in identifying the transition zone, we believe that CT scan should be the preferred primary investigation in adult population with suspected HD. In a study by Seifeldein et al. correct detection of transitional zone by MDCT was made in 86.4% [3]. Although, histopathology is confirmatory, however, visualization of transition zone with dilated proximal bowel segment and the absence of other identifiable causes, along with history of chronic constipation, should prompt towards the diagnosis of HD. In our study, the transition zone was visualized approx. 2.4 cm from the anal verge, a finding consistent with the diagnosis of USHD [4].

Despite multiple counselling sessions emphasizing its fatal complications, the patient declined surgery due to financial constraints and surgical fear. Following a 9 days hospital admission, the patient received conservative management with enemas, laxatives and rectal washouts before being discharged and scheduled for follow-up care.

Conclusion

The scarcity of documented cases and available literature on adult USHD hinders clinicians and radiologists from reaching a diagnosis. This case report underscores the importance of identifying and comprehending this condition based on clinical and radiological findings to improve patient management.

Patient consent

We hereby confirm that we have obtained written, informed consent from the patient featured in this case report/study for the publication of their personal and medical information and have been fully informed of the potential risks and benefits associated with the publication of their case, including the possibility of identification and any potential impact on their privacy and well-being.

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