Hirschsprung's disease: early diagnosis and long-term outcomes

Meinds, Rob Jelle

CHAPTER 5

Solitary rectal ulcer syndrome as a sign of unrecognized Hirschsprung's disease

Rob J. Meinds[1,§], Hubert P.J. van der Doef[2,§], Frank A.J.A. Bodewes[2], Albertus Timmer[3], Monika Trzpis[4], Paul M.A. Broens[1,4]

1 Department of Surgery, Division of Pediatric Surgery, University Medical Center Groningen
2 Pediatric Gastroenterology and Hepatology, University Medical Center Groningen
3 Department of Pathology and Medical Biology, University Medical Center Groningen
4 Department of Surgery, Anorectal Physiology Laboratory, University Medical Center Groningen

§ These authors contributed equally to this work

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SUMMARY

We report on two cases of late diagnosed Hirschsprung's disease. Both cases presented themselves with recurrent solitary rectal ulcer syndrome and were diagnosed with Hirschsprung's disease at 18 and 14 years, respectively. Consequently, we advocate the importance of considering Hirschsprung's disease in pediatric patients with solitary rectal ulcer syndrome.

INTRODUCTION

The diagnosis of Hirschsprung's disease has improved over the past few years and it is diagnosed at increasingly younger ages. Nevertheless, Hirschsprung's disease is still diagnosed after the first year of life in up to 5% of the patients.[1] Patients with a late diagnosis of Hirschsprung's disease variously present with chronic constipation with intermittent episodes of diarrhea, abdominal distension, acute enterocolitis, and/or sigmoid volvulus.[2,3] To our knowledge, literature is limited on solitary rectal ulcers as a presenting symptom of Hirschsprung's disease, as only one Hirschsprung's patient with solitary rectal ulcer syndrome (SRUS) was described before.[4] Consequently, diagnostic tests for Hirschsprung's disease are not included in the standard diagnostic workup of patients with SRUS.[5]

In this case report we describe the medical histories of two pediatric patients with late diagnoses of Hirschsprung's disease and SRUS to illustrate the importance of including anorectal manometry and rectal suction biopsies in the standard diagnostic workup of pediatric patients with SRUS.

CASE REPORTS

Patient 1

In the first patient, who had congenital megacolon, a stoma was created in the transverse colon shortly after birth. As gastrointestinal passage was good and rectal suction biopsies did not confirm Hirschsprung's disease, the stoma was lifted at the age of 6 months. Throughout his childhood, however, the patient suffered from episodes of iron deficiency anemia, rectal bleeding, and constipation. Repeated colonoscopies showed recurrent SRUS (Figure 1A), whereas biopsies taken during the colonoscopies excluded inflammatory bowel disease. Because the complaints recurred, and because the treatment for SRUS (in the form of beclomethason and mesalazine enemas) only helped briefly, resection of the sigmoid was performed at the age of 18 years. Macroscopically, the sigmoid appeared enlarged and dilated. Subsequent microscopic examination of the resected colon showed a non-specific ulceration consistent with SRUS (Figure 1B) and a surprisingly limited number of ganglion cells in the distal resection margin, matching a transitional zone as seen in Hirschsprung's disease. Additionally, along with pronounced anorectal dyssynergia, anorectal manometry revealed that the rectoanal inhibitory reflex was absent, supporting the diagnosis of Hirschsprung's disease.

Patient 2

The second patient suffered from congenital diarrhea and failure to thrive during infancy, upon which parenteral nutrition was initiated. At the time, extensive diagnostic workup (including electron microscopy of duodenal biopsies) failed to reveal the cause of these problems. Eventually, the diarrhea resolved, while growth retardation remained. From the age of 11 years, the patient presented with episodes of iron deficiency anemia, rectal bleeding, and constipation. Three colonoscopies showed persistent SRUS. During hospitalization, severe constipation was suspected as the colon was not properly cleansed after the standard bowel preparations in three subsequent colonoscopies. Upper gastrointestinal contrast series showed an intestinal malrotation without a midgut volvulus while, remarkably, a previous contrast series at the age of 3 months had not shown any evidence of intestinal malrotation. Additionally, at the age of 14 years, a colonoscopy with intensive bowel preparation showed a dilated colon and persistent SRUS.

Due to the similarity between the two patients, we suspected that the second patient might also suffer from undiagnosed Hirschsprung's disease. Indeed, rectal suction biopsies showed aganglionosis and proliferation of acetylcholinesterase positive nerve fibers, confirming the diagnosis of Hirschsprung's disease. Moreover, anorectal manometry revealed an absence of the rectoanal inhibitory reflex, supporting the diagnosis (Figure 2).

A Duhamel procedure at the age of 15 years showed an extremely dilated colon. A fifty centimeters length of colon was resected and the patient received a temporary ileostomy. Furthermore, the intestinal malrotation was corrected. Agenesis of the vermiform appendix was also diagnosed, even though the patient had not previously undergone abdominal surgery.

DISCUSSION

In this case report we describe two pediatric patients who suffered from SRUS, constipation, and chronic anemia, and who were finally diagnosed with Hirschsprung's disease at the age of 18 and 14 years, respectively. In the first patient, the diagnosis was only made after resection of the rectosigmoid which, in retrospect, was inadequate for Hirschsprung's disease. The aim of this case report is, therefore, to draw attention on pediatric patients presenting with SRUS as a possible sign of underlying, misdiagnosed, Hirschsprung's disease. Early detection and proper diagnosis is vital to prevent inadequate treatment strategies.

In pediatric patients, SRUS is an extremely rare and benign disorder. Its clinical

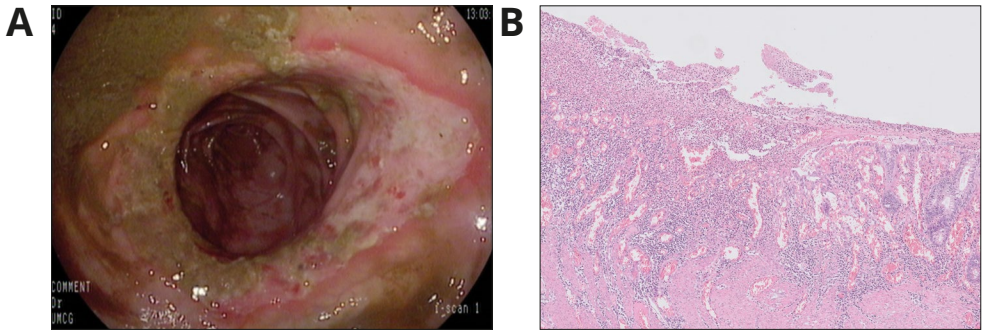

Figure 1
A: Endoscopic findings of a solitary rectal ulcer at the age of 15 years in the first patient.
B: Histological examination of the ulcer after resection of the sigmoid showing findings consistent with SRUS. On the left side non-specific ulceration with granulation tissue and a hyperplastic muscularis mucosae is shown. On the right side preexisting mucosa with erosion, mild inflammation, crypt distortion, and vascular congestion is shown.

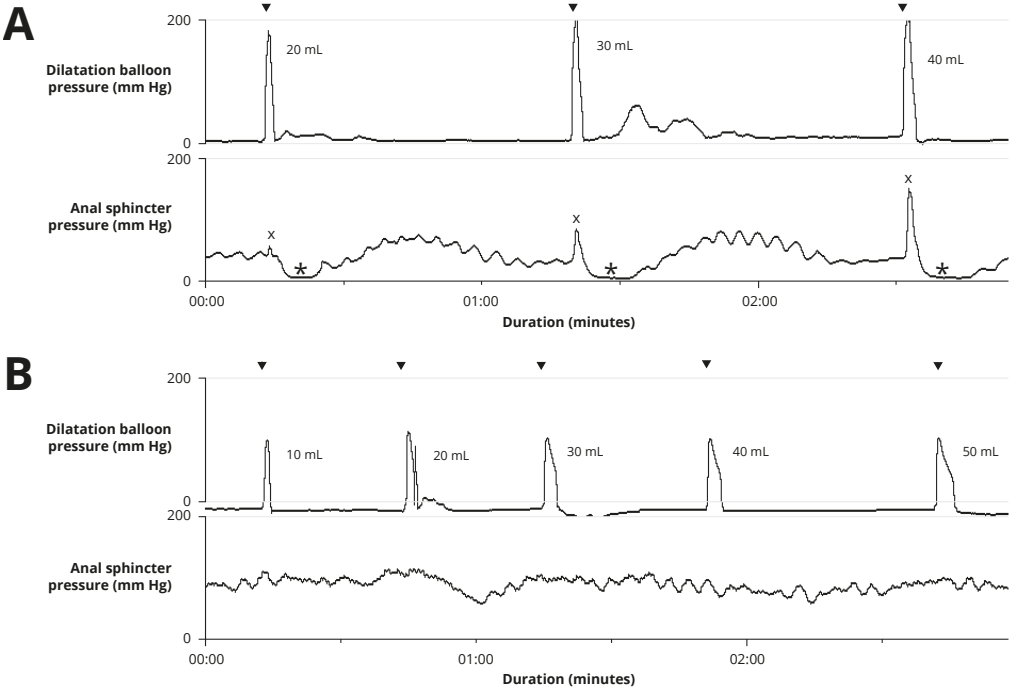

Figure 2
Anorectal manometry measurements showing the presence of the rectoanal inhibitory reflex (denoted by asterisks) in a control subject without Hirschsprung's disease (A), and the absence of the rectoanal inhibitory reflex in the second patient (B). Arrowheads denote the moment of rectal balloon dilation, crosses denote the rectoanal contractile reflex.

presentation is variable, thus complicating the diagnosis.[6,7] Although the etiology of SRUS is incompletely understood, it is thought that local ischemia, and mucosal prolapse as a result of increased rectal pressure, are involved in the pathogenesis of SRUS.[5,8] These factors are also seen in patients with Hirschsprung's disease.[9] While the exact causes of SRUS in the patients described in this report remain unclear, it is possible that an increase of rectal pressure, due to the pathogenesis of Hirschsprung's disease, led to the genesis of SRUS.

To our knowledge, a rectal ulcer in Hirschsprung's disease is mentioned only once in the literature.[4] The presentation of two patients in a relatively short space of time, however, led us to believe that possibly more cases presenting with SRUS are unrecognized cases of Hirschsprung's disease. Further investigations are required to confirm whether at older ages SRUS could indeed be considered as a sign of Hirschsprung's disease.

Previously, it was recommended to examine all pediatric patients with SRUS using defecography and anorectal manometry in order to define an underlying pathophysiological cause.[10] We agree with this suggestion, and in addition, if anorectal manometry cannot exclude Hirschsprung's disease, we strongly advocate performing rectal suction biopsies in any pediatric patient presenting with chronic defecation complaints and blood loss due to SRUS. Adequate treatment for Hirschsprung's disease, even at older ages, might still lead to satisfactory results and improvement of patients' quality of life and could prevent long-term complaints such as growth deprivation and malnutrition.[3]

Basing ourselves on the medical history of these two patients, we conclude that SRUS should be considered as a possible presenting symptom of long-standing, and unrecognized, Hirschsprung's disease. We emphasize the importance of anorectal manometry and rectal suction biopsies as part of the standard diagnostics of pediatric patients with SRUS.

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PART II

Defecation disorders in the general population