Solitary rectal ulcer syndrome and enterocele in a 13-year-old boy

Zespół samotnego wrzodu odbytnicy i enterocele u 13-letniego chłopca

Małgorzata Sładek[1], Stanisław Pieczarkowski[1], Łukasz Wyrobek[2], Krzysztof Fyderek[1]

[1]Department of Pediatrics, Gastroenterology and Nutrition, Jagiellonian University Medical College, Krakow, Poland
[2]Diagnostic Radiology Unit, University Children's Hospital, Krakow, Poland

Prz Gastroenterol 2013; 8 (3): 197–201
DOI: 10.5114/pg.2013.36335

Key words: solitary rectal ulcer syndrome, enterocele, children.
Słowa kluczowe: wrzód samotny odbytnicy, enterocele, dzieci.

Address for correspondence: Małgorzata Sładek MD, PhD, Department of Pediatrics, Gastroenterology and Nutrition, Jagiellonian University Medical College, 265 Wielicka St, 30-633 Krakow, Poland, phone: +48 12 657 40 12, fax: +48 12 658 10 88, e-mail: misladek@cyf-kr.edu.pl

Abstract

Solitary rectal ulcer syndrome (SRUS) is a rare disorder of childhood, which usually presents with any of the manifestations of anorectal disease, bowel habit alteration and abdominal pain. It is commonly accepted that pathogenesis is associated with disturbances of defecation. We describe a 13-year-old boy with attention deficit hyperactivity disorder (ADHD) and SRUS. He complained of increased stool frequency associated with the passage of blood and mucus, tenesmus, sensation of incomplete defecation and abdominal pain. Circumferential ulcer and flat lesion extending up to the sigmoid colon with typical microscopic findings were revealed upon colonoscopy. Defecography showed an opacified small bowel invaginating into the rectal wall, the pictures consistent with enterocele. Conservative treatment, including reassuring the patient and his mother of the benign nature of the disease, recommendation to avoid straining defecation and use of a high-fiber diet to prevent constipation, was applied. The macroscopic appearance of rectal lesions in SRUS are much more variable than is usually realized. An early diagnosis requires a high index of suspicion both for clinicians and the pathologist.

Streszczenie

Zespół samotnego wrzodu odbytnicy (*solitary rectal ulcer syndrome* – SURS) jest rzadkim schorzeniem u dzieci i młodzieży. Zwykle objawia się zaburzeniami oddawania stolca i bólami brzucha. Powszechnie uważa się, że w patogenezie tego schorzenia odgrywają rolę zaburzenia defekacji. W niniejszej pracy przedstawiono opis przypadku 13-letniego chłopca z zespołem nadpobudliwości psychoruchowej i SURS. Głównymi objawami, które zgłaszał pacjent, były: zwiększona częstość oddawania stolca z obecnością śluzu, okresowe krwawienia z odbytnicy, uczucie parcia na stolec i niepełnego wypróżnienia oraz bóle brzucha. W badaniu kolonoskopowym stwierdzono okrężne owrzodzenie i uniesioną, nieregularną zmianę sięgającą esicy z typowym dla SURS obrazem mikroskopowym. Defekografia uwidoczniła typowy dla enterocele obraz wpuklenia się pętli jelita cienkiego w ścianę prostnicy. Chłopcu i jego matce wyjaśniono łagodny, chociaż przewlekły, charakter schorzenia. Leczenie zachowawcze obejmowało zalecenie unikania wysiłkowego oddawania stolca oraz stosowanie diety bogatobłonnikowej, aby zapobiegać zaparciom. W SURS obraz zmian makroskopowych jest bardziej różnorodny, niż powszechnie się uważa. Ustalenie właściwego rozpoznania zależy w dużej mierze od wnikliwości zarówno klinicysty, jak i patologa.

Introduction

Solitary rectal ulcer syndrome (SRUS) is the term used to describe a wide spectrum of abnormalities affecting mainly young adults with varied clinical presentation, protean endoscopic appearance, yet characteristic histopathological findings. In children this condition appears to be infrequent, underdiagnosed or in some cases misdiagnosed [1]. Most patients with SRUS, both adults and children, present with rectal bleeding, mucous discharge, straining, tenesmus, and rectal pain. Accurate diagnosis and treatment of SRUS still remains

a clinical challenge for several reasons. First, the term SRUS is misleading because the lesions may be neither solitary, nor ulcerated, and may even extend beyond the rectum into the sigmoid colon [2]. The macroscopic appearance varies from flat to polypoid or ulcerative lesions, the latter being the most frequently identified. Second, patients present a wide spectrum of clinical signs and symptoms which may mimic other rectal pathologies such as neoplasm or inflammatory bowel disease [3]. Third, lack of sufficient experience may cause difficulty for the clinicians to distinguish it from other disorders. Finally, the pathogenesis remains enigmatic, with evidence that it is multifactorial and associated with abnormal defecation. A typical histology with the presence of fibromuscular obliteration of the lamina propria and disorientation of muscle fibers is crucial for a diagnosis [3]. The pediatric experience with this condition is limited and there is a lack of diagnostic and therapeutic protocols [1, 4-6]. It will be of benefit for pediatric gastrologists to familiarize themselves with this entity, so that patients can be diagnosed correctly before long duration of symptoms. Awareness of the disease may result in more diagnosed cases and help better understanding of the natural history.

We describe a 13-year-old boy with SRUS, who was diagnosed two months after onset of symptoms.

Case report

A 13-year-old boy was referred to our institution after one-month history of increased stool frequency occurring mainly during daytime (6-12 daily) associated with passage of blood and mucus, and weight loss of 2.5 kg in the last month. He complained of central and right quadrant abdominal pain, tenesmus, and sensation of incomplete defecation and decreased appetite. His medical history was significant for attention deficit hyperactivity disorder (ADHD) under psychiatric non-pharmacological treatment. There was no history of inflammatory bowel disease or similar symptoms in other family members. At the time of his initial evaluation, his anthropometric measurements were within normal values both for height and weight. On the physical examination there was mild discomfort on deep palpation in the right abdominal quadrant. Inspection of the perianal area revealed no abnormality except for patulous anus. Digital rectal examination was unremarkable. Routine hematologic and biochemical parameters and coagulation profile were within normal ranges. His erythrocyte sedimentation rate was 16 mm in 1 h, C-reactive protein was < 3 mg/l. Serologic markers specific for inflammatory bowel disease (perinuclear antineutrophil cytoplasmic antibody and anti-*Saccharomyces cerevisiae* antibody) were undetectable. Stool cultures were negative for pathogenic microorganisms (*Salmonella*, *Shigella*, *Yersinia enterocolitica*, *Campylobacter* and pathogenic *Escherichia coli*), ova, parasites and *Clostridium difficile* toxins. Endoscopic examination was performed to address the possibility of inflammatory bowel disease. Upper GI endoscopy showed fine nodular deformity of the antrum with a positive rapid urea test. Histology with hematoxylin-eosin and Giemsa stain confirmed *Helicobacter pylori* gastritis. Colonoscopy up to the cecum was normal except for the rectal findings. A circumferential ulcer 3 to 4 cm in length 6 to 7 cm distal to the anal verge with the surface covered with fibrinous exudates was revealed (Figure 1). The ulcer was well demarcated from the surrounding hyperemic mucosa. Several biopsies were taken from the margin of the ulcer, surrounding mucosa and from normal looking parts of the total colonic segments. Histopathological examination of the mucosa from the lesion showed granulation tissue. Biopsy samples taken from normal looking mucosa were normal on histology. The patient was treated for presumed inflammatory bowel disease with topical 5-ASA with mild improvement. Because of persistence of clinical symptoms, ambiguous histology and the patient's unusual behavior associated with defecation (he used an enormous amount of toilet paper), and ADHD causing difficulty to either report or describe symptoms, repeated colonoscopy was performed. Similar circumferential ulceration in the rectum and additionally an elevated lesion (10 mm × 5 mm) of irregular surface and easy friability in the rectal/sigmoid transitional area were found. The second biopsy set showed ulceration with inflammatory cell infiltration and changes consistent with SRUS including fibrous obliteration of the lamina propria with disorientation of the muscularis mucosa and extension of muscle fibers into the lamina propria (Figure 2). Anorectal manometry demonstrated slightly low resting and squeezing anal canal pressure. Defecography was performed to investigate the anorectal function and showed the opacified small bowel invaginating into the rectal wall, the pictures consistent with enterocele (Figures 3 A and B). Putting together the clinical symptoms, results of macroscopic and microscopic examination, and defecography, the diagnosis of solitary rectal ulcer syndrome was finally established. The standard eradication treatment for *Helicobacter pylori*, topical 5-ASA, stool softeners and defecation training were introduced. The child and his mother were reassured of the benign nature of the disease. Instructions including avoidance of straining and use of a high-fiber diet to prevent constipation were given.

Discussion

Solitary rectal ulcer syndrome is a chronic, benign disorder of the rectum, occurring most commonly in young adults [3]. This entity is either rare in children, goes unrec-

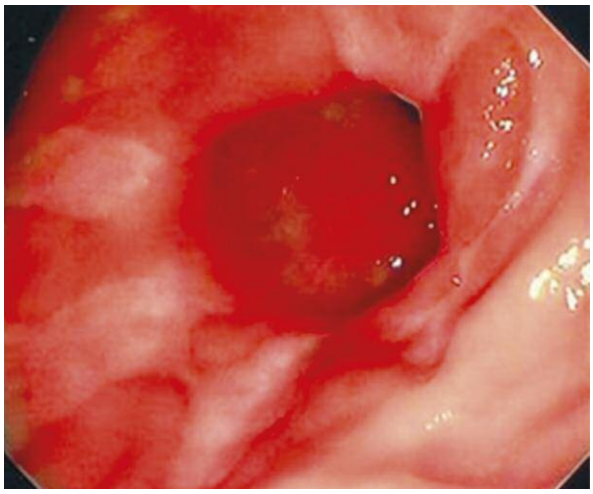

Fig. 1. Colonoscopic examination. A circumferential ulcer in rectum
Ryc. 1. *Kolonoskopia. Okrężne owrzodzenie w odbytnicy*

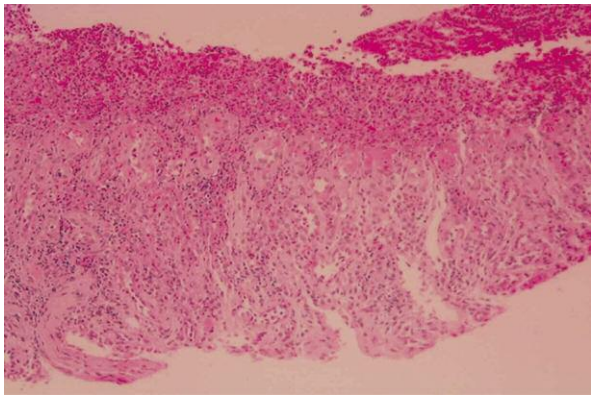

Fig. 2. Histopathology section of the rectal mucosa showing inflamed mucosa and fibromuscular obliteration of the lamina propria (HE stain)
Ryc. 2. *Badanie histopatologiczne bioptatu śluzówki prostnicy. Naciek zapalny z blaszką mięśniową obliterującą w blaszkę właściwą błony śluzowej jelita (barwienie hematoksylina + eozyna)*

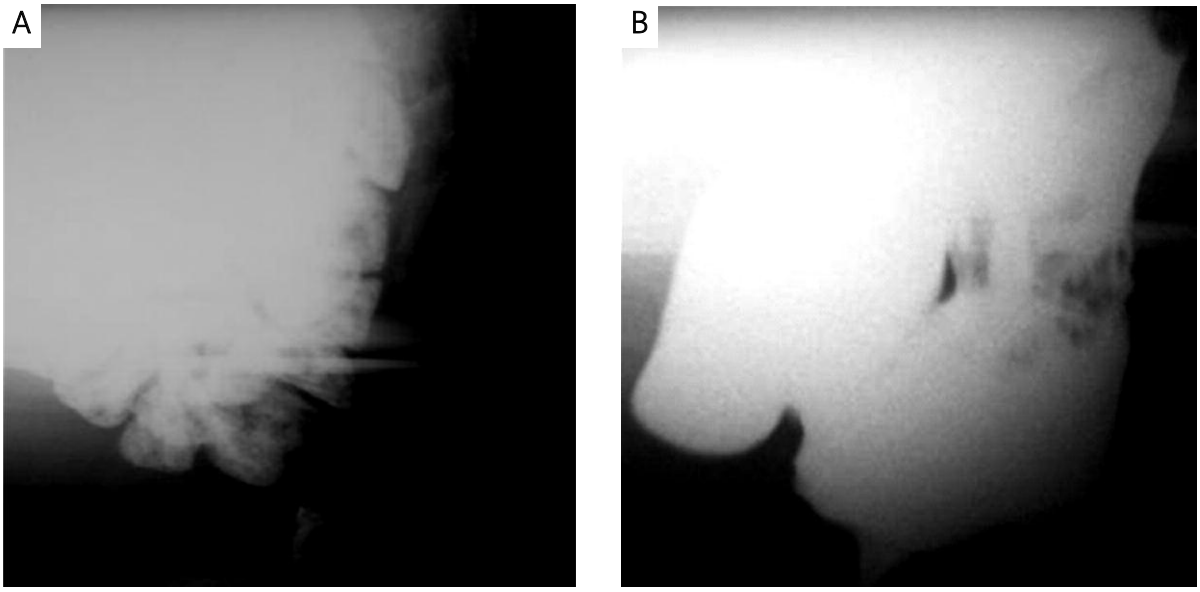

Fig. 3. Defecography (**A**) showing opacified small bowel invagination into the rectal wall during straining (**B**)
Ryc. 3. *Defekografia (**A**) ukazująca wpuklanie się pętli jelita cienkiego w ścianę górną odbytnicy (**B**)*

ognized or is misdiagnosed, with slightly more than twenty documented cases in the English literature [1]. The presented patient illustrates the importance of detailed investigation in children with a rectal endoscopic lesion, passing blood and mucus, and symptoms associated with defecation. To our knowledge this is the first case of documented enterocele in a pediatric SRUS patient.

A typical SRUS appears as isolated erythema or a shallow and solitary ulcerating lesion highlighted by erythematous mucosa, usually located on the anterior rectal wall, several centimeters from the anal verge [7]. When it occurs in a classic form, SRUS can be easily recognized if endoscopists or clinicians keep this disease in mind. However, as mentioned above, the term "solitary rectal ulcer" is misleading. Even in adults, misdiagnoses have been reported in one fourth of cases and the diagnosis was usually delayed, ranging from 3 months to 30 years (mean 7 years) [3, 7]. There are many reasons for SRUS to escape prompt and accurate diagnosis. One important factor may be unfamiliarity of clinicians with

the SRUS macroscopic appearance of rectal lesions, which are much more variable than is usually realized. In general, endoscopic findings can be classified into three types: ulcerative, polypoid, and flat lesions. Ulcers are found in less than half of the patients, but the solitary type is present only in 20 per cent of reported cases. Despite the term solitary, the ulcers may be multiple or circumferential and, like non-ulcerated lesions, vary in shape and size [7]. Even concerning localization, the lesions are not exclusively limited to the rectum, and sigmoid colon involvement has been reported [2]. If one keeps in mind the fact that macroscopic lesions may not be exclusively solitary or ulcerated, misinterpretation of erythema or ulcerated lesions as inflammatory bowel disease and polypoid as neoplasm can be avoided. A histological examination therefore plays a key role in diagnosis as diverse SRUS endoscopic appearance may be misleading. A biopsy should always be taken to make a positive diagnosis and rule out the possibility of other disease. Microscopic findings are highly characteristic and include the presence of fibrous obliteration of the lamina propria with disorientation of the muscularis mucosa and upward extension of smooth muscle fibers into the lamina propria [3]. Careful examination of the rectum and anorectal area, and performing a full ileocolonoscopy examination with multiple biopsy obtained from both pathologic and normal looking sites, are the most important diagnostic investigations of the children with rectal bleeding. Therefore, recognizing the presence of fibromuscular obliteration of the lamina propria is crucial to distinguish it from inflammatory bowel disease, the most common diagnostic confusion in children.

Most patients with SRUS present without any manifestation of anorectal disease, bowel habit alteration, and even abdominal symptoms. Rectal bleeding, mucus discharge, excessive straining, and rectal pain are consistent but not specific symptoms. Other common complaints are lower abdominal pain, increased frequency of defecation, self digitations, and fecal incontinence [3]. A disturbance in bowel habits was reported as constipation or diarrhea. A thorough history taking is of utmost importance in the initial diagnosis. Clinical evaluation of bowel habits is sometimes difficult but it is important to carefully interpret the patient's complaints of diarrhea. Most complaints of diarrhea arise because of frequent passage of mucus in response to tenesmus and dyschezia. The feeling of incomplete evacuation necessitates repeated visits to the toilet and may be misinterpreted as diarrhea by patients. Therefore, clinicians should pay much attention to these complaints to avoid misinterpretation.

In children, the reported prevalence rate for rectal bleeding, mucus discharge and constipation was similar to adults, while rectal pain and self digitations were not reported commonly [1, 3, 4].

Anorectal physiological studies performed in patients with SRUS have given inconsistent results. These may be explained at least partly by taking into count the fact that pathogenesis of SRUS may involve both sphincter outlet obstruction and internal procidentia of the rectal wall. Anorectal pressure may be normal but in some patients reduced resting and squeeze pressure were documented. Half of adult patients with SRUS demonstrated decreased anal tone and one third of them on rectal examination showed a patulous anus [8]. Therefore, anorectal manometry provides additional information in identifying sphincter abnormality in patients with symptoms associated with defecation [9]. Defecography has rarely been performed in pediatric cases and usually is reserved for the investigation of the underlying physiology. It may be a helpful tool to identify the underlying pelvic floor dysfunction [10]. In adults with histologically proven SRUS unequivocal abnormalities on defecography were demonstrated. Internal or external rectal prolapse has been reported most frequently [3, 8]. Only a few cases of enterocele have been documented in adults [11].

The etiology of SRUS still remains not fully understood. It is universally accepted that multiple factors contribute to SRUS development, but the underlying mechanism still remains a subject of debate. Physiological and histopathological studies suggest a spectrum of disease, raising the possibility of a variety of causes in different patients. Disorders of defecation have received the most intensive attention. Several hypotheses have been proposed including lack of relaxation of the puborectalis muscle, inflammation, and localized bowel ischemia. Occult or overt rectal prolapse and paradoxical contraction of the pelvic floor muscles are the most commonly attributed factors involved in development of SRUS. In the present case chronically increased intraabdominal pressure associated with straining and relaxation of the levator ani may have caused the small bowel to be forced into the rectal wall, as a consequence causing rectal wall congestion, edema and ulceration.

Psychological problems associated with the syndrome have been explored only superficially. A disturbance of toileting behavior as an expression of psychological problems appears to be an important pathogenic factor in some patients [3, 12].

Because of the unknown underlying etiology and the limited number of controlled trials, there is no definitive treatment recommendation for SRUS patients. The treatment of SRUS remains problematic and several options have been used in the management of SRUS, ranging from behavioral modification to topical treatment, biofeedback and surgery [4, 8, 10]. Dietary and behavioral modification are recommended in patients with mild to moderate symptoms and in the absence of rec-

tal prolapse [10]. Treatment options mostly depend on severity of symptoms and underlying pathophysiology. The goal of the treatment is to improve disorders of defecation. No consensus exists on what medical or surgical treatment should be used. An individual approach is most likely to succeed. Symptoms may be improved by treatment but it is uncommon to achieve endoscopic and histological normality. Patient education and behavioral therapy remain the cornerstone of treatment of SRUS. Management must include patient reassurance that the underlying lesions are benign and the goals of therapy should be discussed with the patient and the family. Once the diagnosis is established the patient should be instructed on a high-fiber diet, use of laxatives and avoidance of straining. Time spent on the toilet should be minimized, and defecation training emphasized. Solitary rectal ulcer syndrome however may be resistant to conservative treatment. Surgical methods are reserved for patients with SRUS refractory to conservative treatment and biofeedback or in those with significant mucosal prolapse [8, 10].

Conclusions

Because the clinical and macroscopic presentation varies, an early diagnosis requires a high index of suspicion both for the clinician and for the pathologist to consider SRUS in differential diagnosis. Solitary rectal ulcer syndrome should be considered only in the differential diagnosis of idiopathic inflammatory bowel disease limited to the rectum and distal sigmoid colon, but not in the differential diagnosis of more extensive disease. A report of a larger series with long-term follow-up is required to establish treatment protocols in children.

References

1. Ertem D, Acar Y, Karaa EK, et al. A rare and often unrecognized cause of hematochezia and tenesmus in childhood: solitary rectal ulcer syndrome. Pediatrics 2002; 10: e79.
2. Mackle EJ, Manton-Mills JO, Parks TG. The investigation of anorectal dysfunction in the solitary rectal ulcer syndrome. Int J Colorectal Dis 1990; 5: 21-4.
3. Figueroa-Colon R, Younoszai MK, Mitros FA. Solitary ulcer syndrome of the rectum in children. J Pediatr Gastroenterol Nutr 1989; 8: 408-12.
4. Iwańczak F, Stawarski A. Wrzód samotny odbytnicy u dzieci. Nowa Pediatria 2002; 30: 189-91.
5. Godbole P, Botterill I, Newell SJ, et al. Solitary rectal ulcer syndrome in children. J R Coll Surg 2000; 45: 411-4.
6. Vaizey CJ, van den Bogaerde JB, Emmanuel AV, et al. Solitary rectal ulcer syndrome. Br J Surg 1998; 85: 1617-23.
7. Chiang JM, Changchien CR, Chen JR. Solitary rectal ulcer syndrome. An endoscopic and histological presentation and literature review. Int J Colorectal Dis 2005; 21: 348-56.
8. Sharara A, Azar C, Amr S, et al. Solitary rectal ulcer syndrome: endoscopic spectrum and review of the literature. Gastrointest Endosc 2005; 63: 755-62.
9. Morio O, Meurette G, Desfourneaux V, at al. Anorectal physiology in solitary ulcer syndrome: a case-matched series. Dis Colon Rectum 2005; 48: 1917-22.
10. Bishop PR, Nowicki MJ. Nonsurgical therapy for solitary rectal ulcer syndrome. Curr Treat Options Gastroenterol 2002; 5: 215-23.
11. Cronje HS, Beer JA, Bam RH. The pathophysiology of an enterocele and its management. J Obstetrics Gyn 2004; 24: 408-13.
12. Jorge N, Habr-Gama A, Wexner SD. Clinical application and techniques of cinedefecography. Am J Surg 2001; 182: 93-101.