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2444-3824/

Solitary rectal ulcer in a teenage patient☆

Úlcera rectal solitaria en paciente adolescente

Solitary rectal ulcer syndrome (SRUS) is an exceptional disorder in paediatric and adolescent patients. It is often confused with other conditions, such as inflammatory bowel disease (IBD), delaying its diagnosis for years after the first consultation. We describe the case of a teenager with symptoms of tenesmus and rectal bleeding initially classified as ulcerative proctitis but who was later, in view of refractoriness to treatment, diagnosed with SRUS.

The patient was a 15-year-old boy with a history of bronchial asthma and a 1-year history of diarrhoea with up to 7 small-volume stools daily with mucus and haematochezia associated with colicky abdominal pain. Laboratory tests were normal. Colonoscopy revealed an erythematous inflamed erosive ring in the rectum with undamaged distal rectal mucosa and rest of the colonic mucosa normal; histological study reported non-specific chronic rectitis. With a presumed diagnosis of ulcerative proctitis, he initiated treatment with topical mesalazine and was referred to the IBD clinic for follow-up.

The patient was shy and withdrawn and reluctant to provide any information, so his mother described the symptoms. Due to the persistence of symptoms despite the treatment initially described, he was treated with a combination of both topical mesalazine with topical budesonide, and topical mesalazine with oral mesalazine, also with no response. Oral corticosteroids were also tried, similarly with no improvement observed.

At that time, in view of the refractoriness to treatment and the non-typical appearance of the ulcerative proctitis in the initial colonoscopy, we reconsidered the diagnosis and decided to repeat the endoscopic and histological study, observing persistence of a circumferential ring around 7–8 cm from the anal margin, polypoid, erosive and friable, but not affecting the distal rectal mucosa (Fig. 1). Biopsies reported benign border and base of the ulcer with development of granulation tissue, regenerative epithelial changes, and fibromuscular proliferation in the lamina propria, with no changes indicative of IBD, suggesting the possibility of a lesion associated with prolapse.

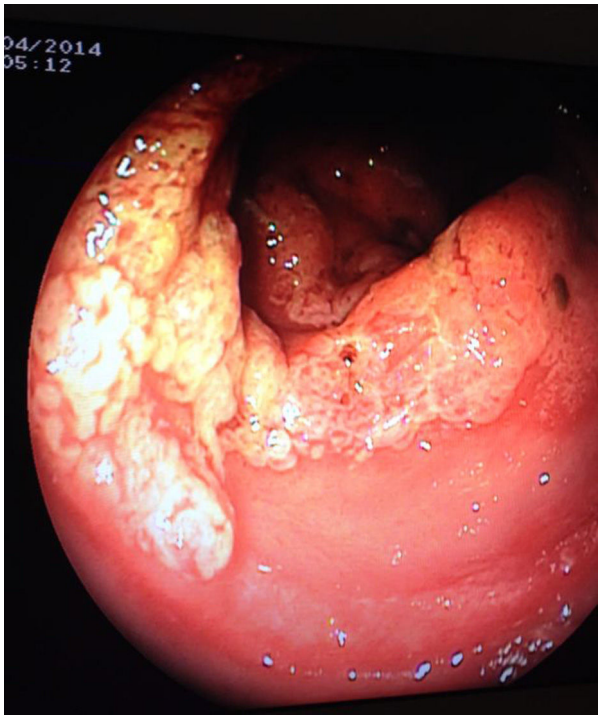

Figure 1 Rectoscopy: circumferential ring in mid-rectum with an erosive polypoid appearance.

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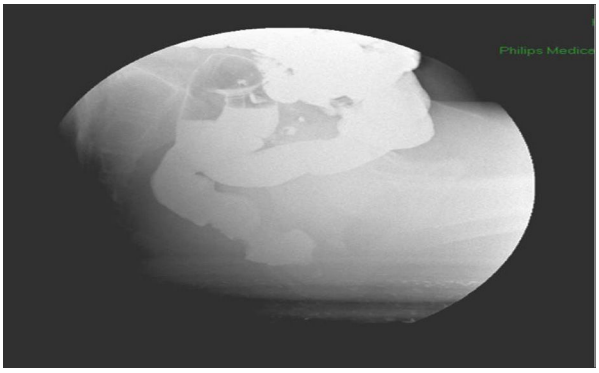

Figure 2 Defecography: internal rectal intussusception.

After re-questioning the patient about the symptoms, with the aforementioned difficulties, the symptoms were more suggestive of tenesmus and defaecation urgency with small-volume stools and incomplete evacuation, all associated with colicky abdominal pain that improved after defaecation, suggesting the possibility of an alteration in rectal defaecation dynamics that in turn led to the onset of SRUS. Video defecography was performed, observing recto-rectal intussusception during defaecation with no anal prolapse (Fig. 2). Anorectal manometry found slight hypotony of the anal sphincter, slight loss of strength in the sphincter muscles and early defaecation urgency with normal expulsion test.

Plantago ovata was prescribed and biofeedback initiated, with a good clinical response to date.

The first clinical and histopathological description of solitary rectal ulcer, a benign chronic disease, dates from 1969.[1] It is a rare entity with an incidence of 1/100 000 population/year, more common in young adults aged between 20 and 35 years, especially in females.[2] Presentation in childhood or adolescence is exceptional, with only isolated case series having been reported.[3–6]

Clinically, it manifests with rectal bleeding, which is usually mild, often accompanied by mucus, as well as tenesmus, straining, feeling of incomplete evacuation, abdominal pain, proctalgia and, occasionally, defaecation urgency.[2–6] There may be some degree of rectal prolapse or internal rectal intussusception accompanied by a feeling of obstruction, although external rectal prolapse has rarely been observed.[6] It has been described as a basic disorder of defaecation dynamics with prolapse of the rectal mucosa through the contracting puborectalis muscle, due to the increased intra-abdominal pressure with consequent strangulation of the rectal mucosa leading to ischaemia, congestion, oedema and ulceration of the mucosa.[7] Another aetiopathogenic possibility is direct trauma as a result of attempts to digitally remove hard stools from the rectum.

Rectoscopy with biopsies is essential for diagnosis. The typical finding is a superficial ulcerated lesion of variable morphology (round, oval, linear or serpiginous) surrounded by an erythematous halo and located in the anterior or antero-lateral wall of the rectum. However, the lesion is not always solitary–it can be multiple or circumferential as in our case–nor ulcerated, with hyperaemic or polypoid lesions also having been described.[8] It can therefore be confused, as in our patient, with other more common entities such as IBD, infectious rectitis, lesions due to sexual abuse or even rectal cancer, with the consequent delay in diagnosis.

The histological findings are characteristic: thickening of the mucosa, elongation and distortion of the glands, oedematous lamina propria with a large amount of collagen and variable proliferation of fibroblasts, and thickening of the muscularis mucosae, with muscle fibres that ascend vertically.[1]

Anorectal manometry and video defecography are useful for assessing the integrity of the rectal sphincters and the physiological state of the anus and rectum, although their diagnostic role is limited.[9]

Treatment of SRUS is problematic, and there are no consensual therapeutic recommendations. It is advisable to reassure the patient and their family that the condition is benign. There are 4 basic pillars of treatment: hygiene-dietary measures, pharmacological treatment, biofeedback and surgery. The response to increasing the intake of dietary fibre combined with bulk-forming laxatives is variable, and patients with associated rectal prolapse are least likely to benefit from these measures. Various topical drugs, such as sucralfate, aminosalicylates and corticosteroids, have been empirically tested with varied results.[6,10] The encouraging results obtained with behavioural therapies in constipation have prompted clinicians to use biofeedback techniques in the treatment of SRUS.[11] One third of patients, especially those who present rectal prolapse, do not respond to the measures described above, and present disabling symptoms that will require surgical treatment, which usually consists of resection of the thickened mucosa and rectopexy.[12]

In short, SRUS is a rare entity, particularly in paediatric and adolescent patients, with a chronic, benign course and which requires a high index of suspicion for early diagnosis.

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Alagille syndrome associated with intestinal atresia☆

Síndrome de Alagille asociado a atresia intestinal

Alagille syndrome (AS) (ALGS; OMIM 118450) has an approximate incidence of 1/30 000 live births.[1] It is a multisystemic disease with an autosomal dominant pattern of inheritance and almost complete penetrance, although around 50–70% of cases are sporadic and caused by *de novo* mutations; furthermore, clinical expression varies greatly. It is a syndrome characterised by hepatic manifestations, mainly cholestasis (96%), butterfly vertebrae (51%), posterior embryotoxon (78%) and congenital heart disease, the most common of which is pulmonary artery stenosis (97%), and peculiar facies (96%). Histopathology study of the liver biopsy shows a paucity of interlobular bile ducts. The defect has been localised in 2 different genes. Mutations in the Jagged1 (JAG1) gene have been identified in more than 90% of cases.[1–3] Prognosis essentially depends on the liver impairment and cardiovascular malformations.[4,5] There is usually some spontaneous improvement in the early years of life, although 15–20% of patients will require liver transplantation.[5,6] This report describes a clinical case of special interest due to the association of AS with intestinal atresia in the neonatal period, reported in very few cases in the literature, and the diagnosis of the child's father as a result of his son's diagnosis, with much less severe expression.

The case involved a full-term newborn, of adequate weight, with pulmonary branch stenosis detected due to heart murmur. Difficulty feeding, vomiting, and abdominal distension with failure to pass the first meconium were obvious from the first hours of life. Suspecting intestinal atresia, abdominal X-ray was performed, revealing absence of rectal gas and distended loops in the upper portion of the abdomen, confirming the diagnosis. Surgery was performed 48 h after birth, with resection of 23 cm of small bowel proximal to the area of stenosis. The infant received parenteral nutrition for 11 days. From day 3, he presented non-isoimmune jaundice with maximum total bilirubin of 12.5 mg/dL. At day 10, a cholestatic pattern of liver enzymes was observed (total bilirubin 6.41 mg/dL, direct bilirubin 4.75 mg/dL, gamma-glutamyl transferase [GGT] 290 U/L, aspartate aminotransferase [AST] 48 U/L, alanine aminotransferase [ALT] 26 U/L), attributed to parenteral nutrition, which was discontinued as a result. After persistence of the cholestasis pattern in follow-up laboratory tests, abdominal ultrasound was performed, in which a full gallbladder was observed, with no bile duct dilatation. Gradual worsening of the cholestasis was noted, with maximum direct bilirubin levels increasing to 10 mg/dL, so the aetiological study was extended. Metabolic study, hormone levels, sweat test, cytomegalovirus testing and alpha-1 antitrypsin were all normal. Magnetic resonance cholangiography (MRC) showed no extrahepatic obstructive signs or obstruction of the main hepatic branches. Hepatobiliar iminodacetic acid (HIDA) scan showed passage of contrast to the duodenum. Liver biopsy found paucity of bile ducts (Fig. 1) which, associated with the cholestasis, heart disease and peculiar facies (Fig. 2), suggested a diagnosis of AS. Ophthalmological

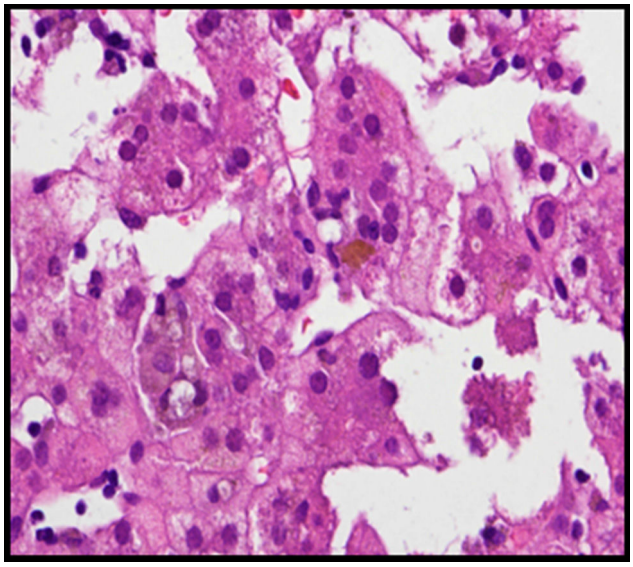

Figure 1 Hepatocytes with presence of intracytoplasmic bile pigment (haematoxylin–eosin).

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