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Enteritis cystica profunda with intestinal intussusception in an infant

Isadora de C. Trevizoli[a,c], Daniele A. Lira[b], Paulo M.S. Lassance[c], Heinrich B.K. Seidler[d], Elisa de Carvalho[a,b,e,*]

[a] Hospital da Criança de Brasília, Brasília, Distrito Federal, Brazil
[b] Centro Universitário de Brasília (UniCEUB), Brasília, Distrito Federal, Brazil
[c] Hospital Materno Infantil de Brasília, Brasília, Distrito Federal, Brazil
[d] Laboratório Brasiliense, Brasília, Distrito Federal, Brazil
[e] Hospital de Base do Distrito Federal, Brasília, Distrito Federal, Brazil

A B S T R A C T

This publication reports enteritis cystica profunda (ECP) in the youngest patient described in the literature, being the fifth case that demonstrates an association between ECP and intestinal intussusception and the seventh case of ECP in children.

A 30-day-old boy presented intense abdominal pain associated with digestive bleeding with “red currant jelly” stool. He had a history of bloody stool at 15 days old. Due to family history of atopy and normal physical examination, allergic proctocolitis was suspected and the patient temporarily improved after cow's milk and dairy products were eliminated from his mother’s diet. Abdominal ultrasound confirmed the hypothesis of intestinal intussusception. During the surgical approach, ileoileal invagination was evident together with a polypoid tumor of the intestinal wall. Resection of the lesion and ileoileal anastomosis was performed. In the postoperative period, he presented intestinal adhesions, with the need of reoperation and, subsequently, dehiscence of the aponeurosis and intestinal perforation. The patient underwent operation with good recovery. He was discharged in good health and maintained satisfactory clinical progress.

The histopathological study of the resected intestine (terminal ileum) revealed a lesion formed by cystic dilations with invasion to the submucosa and musculature of the intestinal wall, lined by intestinal epithelium cells and without nuclear atypia; these findings are compatible with ECP.

1. Introduction

Enteritis cystica profunda (ECP) is a rare disease, with only approximately 20 cases described in the literature. The first description was in 1978 in a 3-month-old child. Since then, 6 cases of ECP in children have been reported, and in 4 of them, the initial presentation was intussusception.

Macroscopically, ECP is characterized by sessile polyps and high lesions, with overlying edematous mucosa, which may present ulcerations; and microscopically, by the presence of submucosal cysts full of mucus in the small intestine [1]. Herein, we report a case of ECP associated with intussusception in a 1-month-old infant, the youngest ever reported patient.

2. Case report

A healthy boy was born via cesarean delivery, weighing 3300 g, length of 48 cm, and head circumference of 33 cm. At 15 days old, he presented with enterorrhagia, accompanied by pallor and weakness. He was exclusively breastfeeding. The consulting pediatrician suspected allergic proctocolitis and indicated a restriction of cow's milk and derivatives to the nursing mother. There was a temporary improvement. The family history revealed that the mother had uveitis, father had allergic rhinitis, and sister and cousins had cow's milk protein allergy (CMPA).

At 30 days old, the infant was brought to the emergency department owing to abdominal pain associated with digestive bleeding and “red currant jelly” stool. Intestinal intussusception was suspected and confirmed on abdominal ultrasonography. Emergency surgery was performed. Surgical findings revealed ileoileal invagination along with a

Abbreviations: ECP, enteritis cystica profunda; CCP, colitis cystica profunda

* Corresponding author. Department of Pediatric Gastroenterology, Hospital da Criança de Brasília José Alencar and Hospital de Base do Distrito Federal, SAIN Lote 4-B, Zip code: 70.071-900, Brasília, Distrito Federal, Brazil.
E-mail addresses: ct.isadora@gmail.com (I.d.C. Trevizoli), elisacarvalho@terra.com.br, elisa.carvalho@hcb.org.br (E. de Carvalho).

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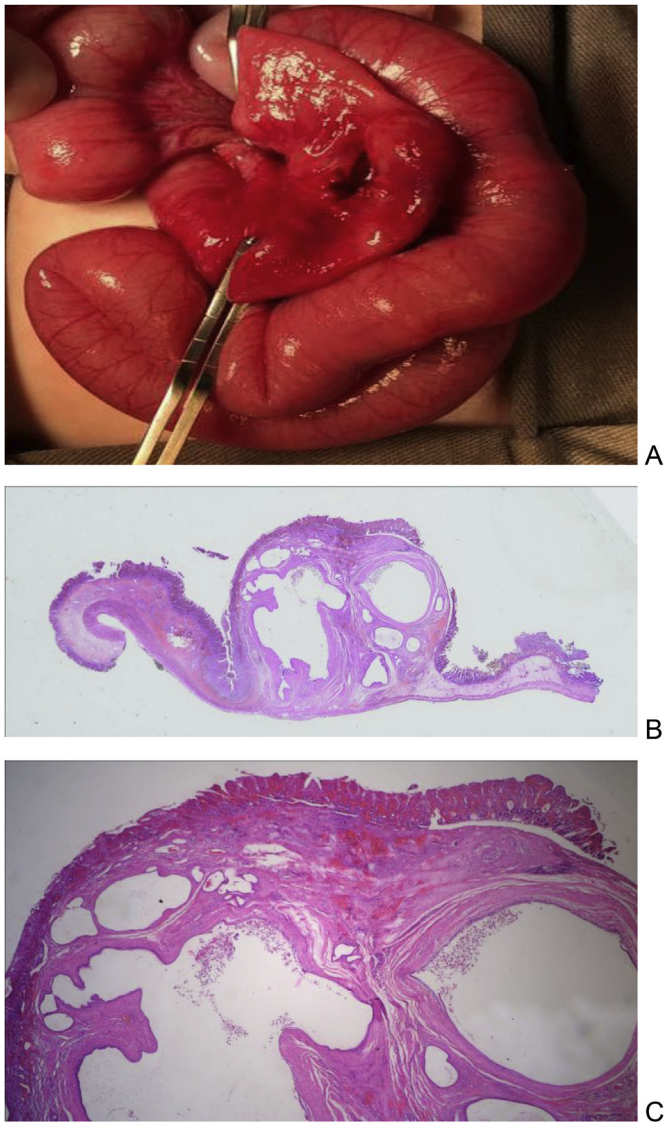

Figure 1. A. Polypoid tumor of the ileum that led to intussusception. **B**. View of the intestinal wall, showing the formation of cystic dilations invading the submucosa and muscularis propria, covered by the intestinal mucosa. The cysts create elevation of the mucosa, by protruding into the lumen, creating a polypoid aspect. **C**. Histology detail, evidencing cysts lined by mitigated epithelium, without atypia. Stroma involving cysts composed of fibroblasts with collagen deposition and proliferation of muscle fibers. The mucosa shows ischemic features.

tumor in the intestinal wall (Fig. 1A). Resection of the lesion, ileoileal anastomosis, and appendectomy were performed. On the fourth postoperative day, the infant presented with abdominal distension and vomiting, compatible with an obstructive presentation. He underwent reoperation, and resection of intestinal adhesions was performed. On the seventh postoperative day, dehiscence of the aponeurosis was noted. At the surgical center, an intestinal perforation was observed near the anastomosis. Subsequently, an ileostomy was performed.

Histopathological study of the resected intestine (terminal ileum) revealed a lesion formed by cystic dilations that invaded the submucosa and musculature of the intestinal wall, lined by reactive intestinal epithelium cells and without nuclear atypia; these findings were compatible with ECP (Fig. 1B). The mucosa covering and adjacent to the lesion showed ischemic traits, possibly owing to a compressive effect, with erosion of the surface epithelium, accompanied by edema, vascular congestion, and mild to moderate mixed inflammatory infiltrate in the lamina propria, without significant eosinophilia, without formation of basal plasmocytosis. The viable epithelium was attenuated, maintaining a regular architectural organization (Fig. 1C).

After ileostomy, patient evolved with high flow on ileostomy and signs of infection on laboratory exams (complete blood count with leukocytosis and left shift, elevation of C-reactive protein and Erythrocyte sedimentation rate). The patient was treated with antibiotics, revealing an improvement in the patient's condition. He was discharged in good health and maintained satisfactory clinical progress.

3. Discussion

This report describes the youngest patient with ECP published to date, being the fifth case that demonstrates an association between ECP and intestinal intussusception and the seventh case of ECP in children.

3.1. Definition

ECP is a very rare lesion of the small intestine, characterized by cystic lesions filled with mucin that penetrate the mucosal muscle and are lodged in the submucosa [2,3]. Profunda cystica lesions are described as a benign entity [4], which may occur throughout the gastrointestinal tract. Cases have been described in the esophagus, stomach, and gallbladder [1,5–7]. The most common site is the colon, being colitis cystica profunda (CCP) a well-defined pathological entity [8], with more than 200 cases described [9].

3.2. History

The first report of the disease was by Stark, in 1766, who described two cases of CCP among seven patients who died of chronic diarrhea [10]. In 1863, Virchow described a case of polypoid lesions containing cysts and coined the term "colitis cystica polyposa" [11]. The first report of involvement of the small intestine was reported in 1970 by Baillie and Abel [12]. The patient was an 11-month-child with multiple congenital cysts in the lungs, kidneys, and the mesentery; who presented intestinal intussusception when polyps were found in the jejunum, which on histology, showed a mixture of small mucus-retention cysts, irregular glandular spaces, and pools of extruded mucin surrounded by chronic productive inflammation of the lamina propria. Four cysts were restricted to the mucosa and one also involved the submucosa.

3.3. Pathophysiology

The pathogenesis of ECP has not yet been fully elucidated, with congenital hypotheses and acquired hypotheses as potential sources. Most of the evidence indicates that the acquired component has a greater influence than the congenital one [13–15]. However, these hypotheses are not mutually exclusive.

One pathogenic mechanism is probably ischemia: an inflammatory stimulus would cause hypertrophy of the wall of the submucosal venules, thereby reducing blood flow and causing local edema and ulceration via focal mucosal necrosis.

The other theory, originally proposed by Goodall and Sinclair [10], assumes that the epithelial cells are found in the submucosa layer after initial ulcerative lesions of the mucosa or inflammatory damage to the muscular layer of the mucosa (usually adjacent to the lymphatic system). This is accomplished through a fissure in the mucosa and migration of cells of the mucosa to the submucosa, leading to cystic dilatation and formation of cysts [16]. Other characteristics that may be found are calcifications, fibrosis, and ossified deposits within mucin lakes [1].

An experimental model in rats emphasizes the role of mucosal ulceration in the development of the ECP; in that study, the small intestine of animals was sutured to the abdominal wall, exposing the mucosa