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Uncommon Pathology in Uncommon Age: Colitis Cystica Profunda in the Youngest Pediatric Case to Date

Golakiya I[1*], Hapani Y[1], Patel R[1] and Vekariya P[2]

[1]Medicine, Surat Municipal Institute of Medical Education and Research, Veer Narmad South Gujarat University, India

[2]Gastroenterology, Jawaharlal Institute of Postgraduate Medical Education & Research, India

*Corresponding author: Golakiya I, Medicine, Surat Municipal Institute of Medical Education and Research, Veer Narmad South Gujarat University, C-1, 104, Krishna Arcade, Ved Road, Dabholi, Surat-395004, India.

Tel: +91-8980053340; E-mail: ishikaagolakiya@gmail.com

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Abstract

Colitis cystica profunda (CCP) is a very rare and poorly understood pathology characterized by the formation of deep colonic cysts. The majority are observed in adults, with only two cases in the pediatric age group documented previously. We present the case of an 8-year-old child, the youngest known patient with CCP reported to date. This case contributes to the limited pediatric literature on CCP and highlights the importance of early diagnosis and tailored management strategies in the pediatric population. Moreover, this case report underscores the importance of proper diagnosis to prevent unnecessary operative interventions and promotes conservative management, which can lead to the best outcomes. Through this presentation, the report underscores the necessity of raising awareness of CCP in pediatric patients and aims to inform clinical decision-making.

Introduction

Colitis cystica profunda is a benign condition that primarily affects the submucosal layer of the colon. Histologically, it is characterized by mucosal cysts that extend deep into the colonic wall, often causing distension of the colonic mucosa. The etiology of CCP is poorly understood, though it has been associated with chronic inflammatory conditions, trauma, or ischemia. While CCP is predominantly reported in adults, mainly those aged 30 Years–60 Years, there have been very few pediatric cases [1–3]. This case shows an extremely rare occurrence of CCP in a minor, making this the youngest patient in available literature to date [1,2,4].

Case Presentation

An 8-year-old male presented to the outpatient department with a history of rectal bleeding and intermittent left lower quadrant abdominal pain over the past three months. The patient had no significant medical or family history of gastrointestinal diseases and no known genetic predisposition. On physical examination, the child appeared well-nourished but reported discomfort on palpation of the lower abdomen. The patient's vital signs were stable. Digital rectal examination findings were unremarkable. Laboratory investigations revealed mild anemia (Hb 10.2 g/dL), and all other parameters were within normal limits.

Abdominal ultrasound did not reveal any apparent structural abnormalities. Given the persistent symptoms and lack of clear etiology, the patient underwent a colonoscopy. The procedure revealed a 4 cm pedunculated polyp in the sigmoid colon. The polyp was removed via detachable end loop and snare polypectomy. The post-procedure inspection confirmed no remaining lesions and active bleeding [3,5,6]. Histopathological evaluation revealed

submucosal cysts lined by benign epithelium, confirming the CCP diagnosis.

Discussion

CCP is one of the rarest benign pathological conditions, characterized by mucin-filled cysts underneath the muscularis mucosa. It was first described in the 18th century by Stark and later named by Virchow. Histologically, CCP is defined by the presence of mucus-filled cysts within the submucosa, which often mimic malignant lesions like mucinous adenocarcinomas [1,2,6]. Most cases occur in the rectum and sigmoid, and CCP is divided into localized and diffuse types. The localized form usually presents as a solitary polyp, while the diffuse form usually occurs in an inflammatory bowel condition [2,4]. While most cases occur in adults, pediatric cases are exceedingly rare, presenting diagnostic challenges. This report is about a very rare pediatric case, along with the diagnostic and therapeutic features of CCP.

The symptoms vary from rectal bleeding, mucorrhea, diarrhea, and colonic obstruction to, indeed, a pure incidental finding at imaging or colonoscopy.

The pathogenesis of CCP remains a debated issue, while it is traditionally based on a combination of congenital muscularis mucosa weakness, continuous inflammation, ischemia, or trauma causing epithelial herniation. Histopathology remains critical in the diagnosis, distinguishing the condition from malignancies like mucinous adenocarcinoma through the absence of cellular atypia or invasive features.

Imaging modalities like CT and MRI are considered for differential diagnosis by identifying cystic, non-invasive submucosal lesions. However, the contribution of endoscopy and histopathology is considered indispensable [2,3,6]. Endoscopic findings often include nodular or polypoid lesions with normal or edematous mucosa. In this instance, colonoscopy demonstrated a pedunculated polyp, histologically confirmed as CCP.

Treatment varies according to the size and symptomatology of the lesion. Localized CCP is treated conservatively with diet and lifestyle modifications to reduce straining and constipation, whereas CCP with severe obstructive symptoms and recurrent episodes requires endoscopic removal [1,4,5]. Radical surgical approaches are indicated in cases with complications like obstruction or suspicion of malignant disease. Its importance in cases of recurrence is monitoring through follow-up colonoscopy, but it would generally mean an excellent prognosis with proper management of the condition [7].

Conclusion

This case report presents the youngest pediatric case of Colitis cystic profunda, highlighting the diagnostic challenges and the importance of highlighting rare conditions in the differential diagnosis of gastrointestinal symptoms. CCP is typically found in adults, but it is important to remember that it can be recognized in children, even though it is rare. Early diagnosis through colonoscopy and histopathological examination is key to appropriate management, which in this case involved a conservative approach with positive outcome.

This report expands the clinical understanding of CCP and may encourage further research into its pathogenesis, particularly in pediatric populations.

Conflict of Interest

The authors declare no potential conflicts of interest with respect to the research, authorship, and/or publication of this article. Informed consent was obtained for this publication.

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