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CASE REPORT | COLON

Perisigmoid Abscess Leading to a Diagnosis of Ehlers-Danlos Syndrome Type IV

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Abstract

The Ehlers-Danlos syndromes (EDS) are a group of connective tissue disorders characterized by triad of joint hypermobility, skin extensibility, and tissue fragility. Ehlers-Danlos syndrome type IV places patients at risk for life-threatening, spontaneous, vascular or visceral rupture due to reduced or abnormal secretion of type III collagen. We present an adolescent male who was found to have a perisigmoid abscess with a fistula connecting to adjacent sigmoid colon secondary to undiagnosed EDS type IV. Conservative management with antibiotics and bowel rest was pursued to allow for elective resection for his acute complicated diverticulitis at a safer time.

Introduction

The Ehlers-Danlos syndromes (EDS) are a group of connective tissue disorders characterized by a triad of joint hypermobility, skin extensibility, and tissue fragility. The Villefranche criteria recognize 6 subtypes, based on clinical characteristics, mode of inheritance, biochemical, and molecular findings.[1] Vascular EDS leads to reduced or abnormal secretion of type III collagen and clinically presents with thin, translucent skin, easy bruising, and characteristic facial features. These characteristics are less noticeable than the hyperflexibility of skin and joints that is seen in other types of EDS; thus, diagnosis is often made after complications such as spontaneous vascular or visceral rupture have occurred. Vascular rupture or dissection and gastrointestinal perforation or organ rupture are the presenting signs in 70% of cases, with 80% of individuals experiencing these complications by age 40 years.[2]

Case Report

A 15-year-old adolescent boy with a suspected bleeding disorder presented with a 4-day history of intermittent suprapubic pain, constipation, decreased appetite, and low-grade fevers. He was febrile and tachycardic, with guarding and lower abdominal tenderness on exam. Laboratory studies were significant for elevated C-reactive protein and leukocytosis. Abdominal x-ray and ultrasound were normal. Abdominal computed tomography (CT) demonstrated bowel wall thickening and fat stranding from the sigmoid colon to the rectum consistent with colitis, as well as a small perisigmoid abscess (Figure 1). He was started on intravenous piperacillin-tazobactam with resolution of his symptoms and improvement in inflammatory markers. He was evaluated for possible drainage of the abscess; however, it was deemed too small and the patient was discharged with oral antibiotics.

Three days post-discharge, the patient was readmitted with similar complaints. Repeat CT demonstrated significant interval enlargement of the abscess (Figure 2). The drainage appeared feculent and concerning for fistula. An abscessogram demonstrated a fistula between the abscess and the adjacent sigmoid colon. Sigmoidoscopy demon-

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Figure 1. CT showing a small abscess (arrow) and thickened sigmoid colon wall with adjacent inflammatory changes (asterisk).

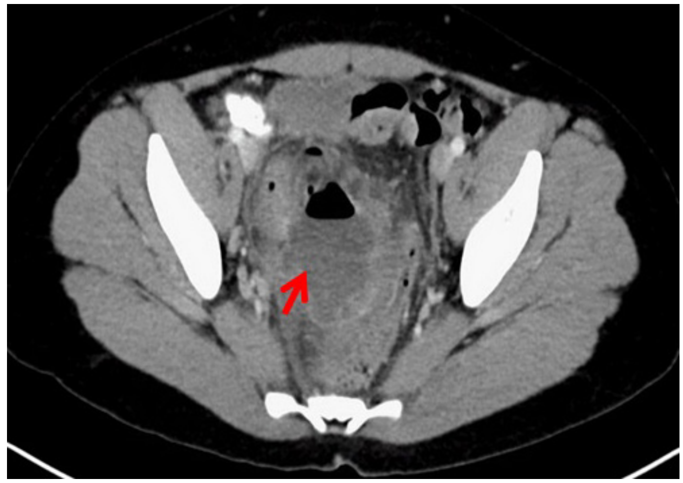

Figure 2. Increased size of the abscess (arrow) with increased surrounding inflammatory changes.

strated cobblestoning and friability in the sigmoid colon with ulcerated lesions in the descending colon and rectum (Figure 3), concerning for Crohn's disease. However, the histological findings were not consistent with Crohn's disease, but rather showed multiple mucosal areas with nonspecific reactive lymphoid nodules with congested and inflamed diverticular-like folds. These findings were compatible with diverticular disease-associated segmental colitis.

Careful review of his medical chart revealed that he presented to his pediatrician years ago with complaints of easy bruising since infancy, including gingival bleeding and frequent epistaxis with a negative work-up. A history of joint hypermobility triggered suspicion for a collagen-vascular disorder, such as EDS. Due to the high suspicion for EDS, a conservative approach to treatment of the sigmoid fistula and abscess was utilized. He remained on antibiotics and was discharged home with the drainage catheter in place.

He was evaluated with another abscessogram post-discharge and was found to have a persistent fistula. At that point, 10 mL of fibrin glue was injected into the abscess in an attempt to seal it and the drain was removed. He had recurrence of his symptoms shortly after, for which he was treated with a course of antibiotics. He was found to have a mutation of the COL3A1 gene, consistent with the diagnosis of vascular EDS. He has remained well without the need for surgery.

Discussion

Diverticulosis, while a common problem in adults, is rare in children. When it is seen in children, it may be due to alterations in the components of the colonic wall as a result of genetic mutations such as EDS, Marfan syndrome, and Williams-Beuren syndrome.[3-6] Diverticulitis in younger patients is considered to be a relative indication for early surgery, as the disease has been described as more aggressive in this age group.[7-9] However, in patients with diverticulitis caused by vascular EDS, non-surgical management with antibiotics and bowel rest should be considered, as these patients are at high risk of complications, including poor hemostasis, tissue friability, formation of extensive intraperitoneal adhesions, fistulas, evisceration, poor wound healing, and wound dehiscence.[10,11] Several authors have concluded that surgery should be avoided in these patients, unless the procedure is considered to be life-saving.[10,12,13]

EDS is a rare disorder with significant surgical and vascular complications. One must have a high index of suspicion for

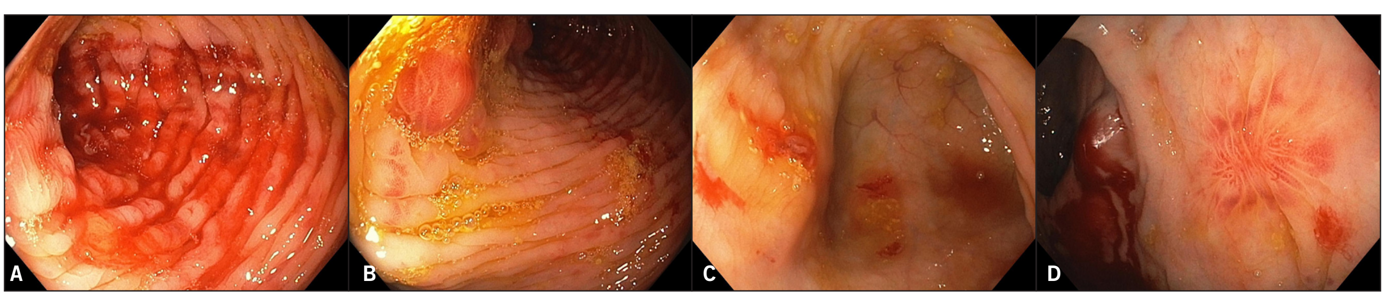

Figure 3. Sigmoidoscopy showing (A) cobblestoning of the sigmoid colon, (B) sigmoid colon ulceration, (C) descending colon with normal proximal mucosa and ulcerated distal descending colon, and (D) ulcers in rectum with adjacent normal-looking mucosa.

EDS when presented with diverticular disease in the pediatric population. We propose that conservative medical management with antibiotics and bowel rest allows for elective resection at a safer and later time in vascular EDS patients with acute complicated diverticulitis.

Disclosures

Author contributions: I. Normatov wrote, edited, and approved the manuscript. A. Kesavan revised the manuscript and is the article guarantor. PB Srikumar and RM McConnie approved the final version of the manuscript.

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