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CASE REPORT | COLON

A Unique Presentation of Familial Idiopathic Colonic Varices

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ABSTRACT

Colonic varices typically occur in the setting of portal hypertension, and patients may present with rectal bleeding or occult anemia. Idiopathic colonic varices occur infrequently in the absence of cirrhosis and can involve the entire colon. We present a case of a 54-year-old Eastern European woman who had undergone diagnostic colonoscopy for newly diagnosed sigmoid adenocarcinoma and was incidentally found to have colonic varices with normal portal pressure gradients. Her 38-year-old daughter was found to have similar varices, raising concerns for hereditary etiology.

KEYWORDS: Ileocolonic varices; hereditary; familial; portal pressure gradient; adenocarcinoma

INTRODUCTION

Colonic varices are defined by the presence of dilated vasculature in the colonic mucosa. They most commonly occur because of portal hypertension associated with hepatic or systemic dysfunction, and diagnosis is typically made by colonoscopy or computed tomography (CT) angiography. Isolated rectal varices occur in 36%–90% of patients with portal hypertension; however, pancolonic varices occur less often in this population.[1–4] Idiopathic colonic varices are not associated with systemic disease and are much rarer, with less than 100 cases reported.[5,6] They are most often confined to the large intestine, and very few cases extend into the small bowel mucosa. Patients are often diagnosed incidentally or because of recurrent bleeding, which can be associated with significant morbidity and adverse health outcomes. Although data are limited because of rarity, idiopathic colonic varices may also have a hereditary association, with up to 33% of cases demonstrating familial involvement.[7] While there have been individual cases with intestinal varices and normal portal pressures, adult familial case series are uncommon.[8,9] We present a mother-daughter case of idiopathic ileocolonic varices with normal portal pressure gradients.

CASE REPORT

A 54-year-old Eastern European woman was referred to our institution to undergo diagnostic colonoscopy for a positive fecal immunochemical test. Her medical history was significant for a remote history of large-volume gastrointestinal bleeding without workup. Index colonoscopy was notable for a nonobstructing sigmoid mass with central depression and serpiginous blue vasculature in the upper rectum extending throughout the colon. There were no stigmata of bleeding. Biopsies were consistent with adenocarcinoma (Figure 1). Workup before segmental colonic resection included liver biopsy, which was notable for steatosis without cirrhosis and a portal pressure gradient of 4 mm Hg. Abdominal magnetic resonance imaging was also completed and notable for persistent colonic varices (Figure 2).

It was discovered that the patient's 38-year-old daughter had previously experienced intermittent rectal bleeding and had undergone endoscopic evaluation 12 years earlier, which was notable for colonic varices with ileal extension. At that time, abdominal CT angiography was negative for evidence of vascular disease or thrombosis, and liver biopsy was unremarkable. On further evaluation, she denied abdominal pain, weight loss, fevers, chills, hematochezia, or melena. She denied a history of alcohol use or illicit substance use. Body mass

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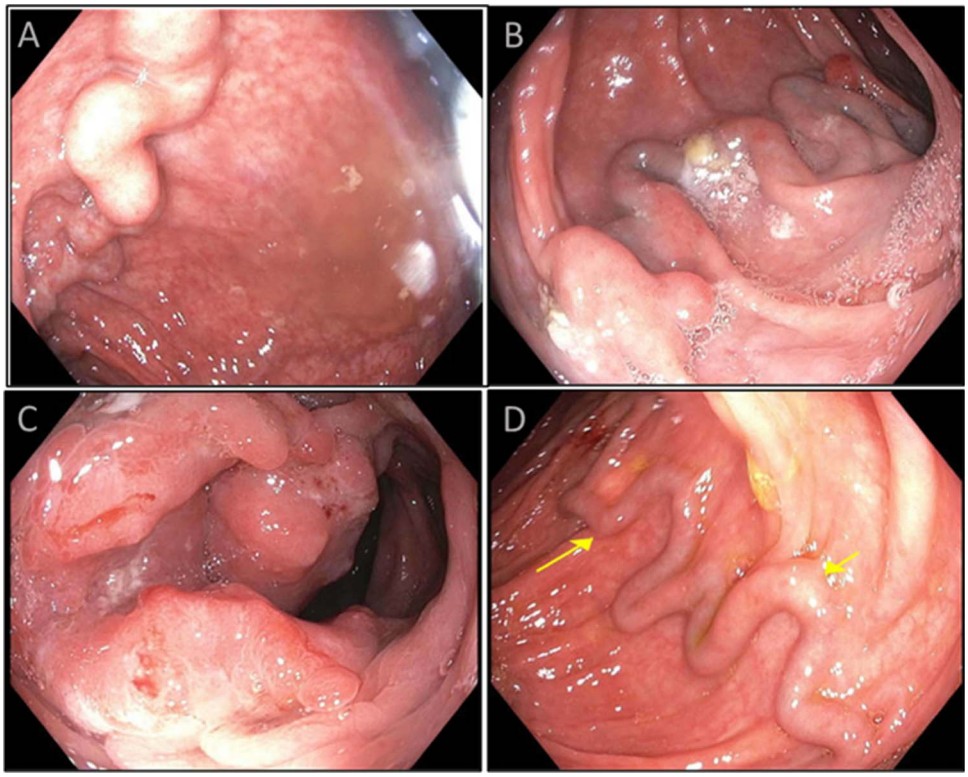

Figure 1. Colonoscopy images of colonic varices and sigmoid mass in a 54-year-old woman. (A) Rectum, (B) sigmoid colon, (C) sigmoid mass, and (D) descending colon.

index was 29. Laboratory testing was notable for hemoglobin 14.1 g/dL, mean corpuscular volume was 93.7 fL, and liver function testing was within normal limits. Abdominal ultrasound was completed with normal liver anatomy and vasculature.

Owing to her new colon malignancy, the patient's daughter underwent screening colonoscopy, which revealed at least columns of large, nonbleeding varices extending from the rectum to the cecum (Figure 3). There was also evidence of variceal extension to the terminal ileal mucosa (Figure 3). A conservative management approach was followed with recommendations for repeat screening colonoscopy in 5 years.

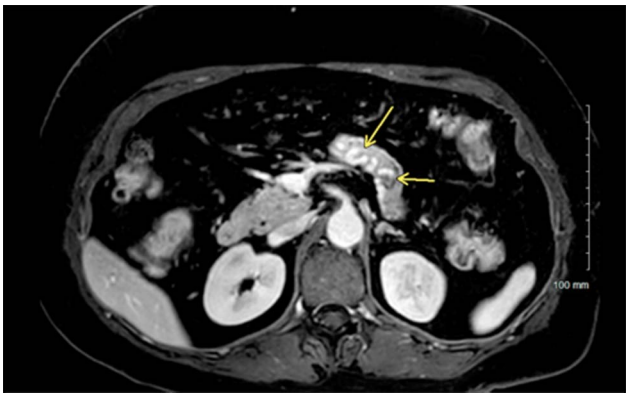

Figure 2. Abdominal magnetic resonance imaging evidence of colonic varices in the 54-year-old woman. Arrows indicate the presence of varices.

DISCUSSION

Colonic varices are a rare condition characterized by vascular dilation in the colonic mucosa. They usually develop with persistently elevated portal pressures in the setting of advanced liver disease or vascular anomalies, such as thrombosis or angioectasias.[4,5,7,10–12]

Varices are often identified during evaluation for acute rectal bleeding or long-standing anemia, or they can be found incidentally on routine screening endoscopy.[5,12] Cases associated with portal hypertension are more likely to present with progressive symptoms and are often identified with CT angiography because of acute decompensation.[13,14] This is associated with significant patient morbidity related to significant bleeding requiring hospitalization and urgent intervention.[14–16] These patients are typically diagnosed at a young age and are more likely to have vascular abnormalities requiring surgical resection.[17] Patients presenting in and beyond the fifth decade can report up to 30 years of preceding bleeding episodes without a diagnosis, which suggests a relapsing and resolving pattern to the varices and portal flow.[7,13]

When systemic pathology is not identified on extensive evaluation, varices are labeled as idiopathic. Recommended diagnostic evaluation is focused on portal pathology and includes abdominal CT angiography, abdominal magnetic resonance imaging, colonoscopy, liver enzyme testing, and possibly liver biopsy. These cases are rare, with less than 100 reported, and they occur most commonly in male patients during the first 50

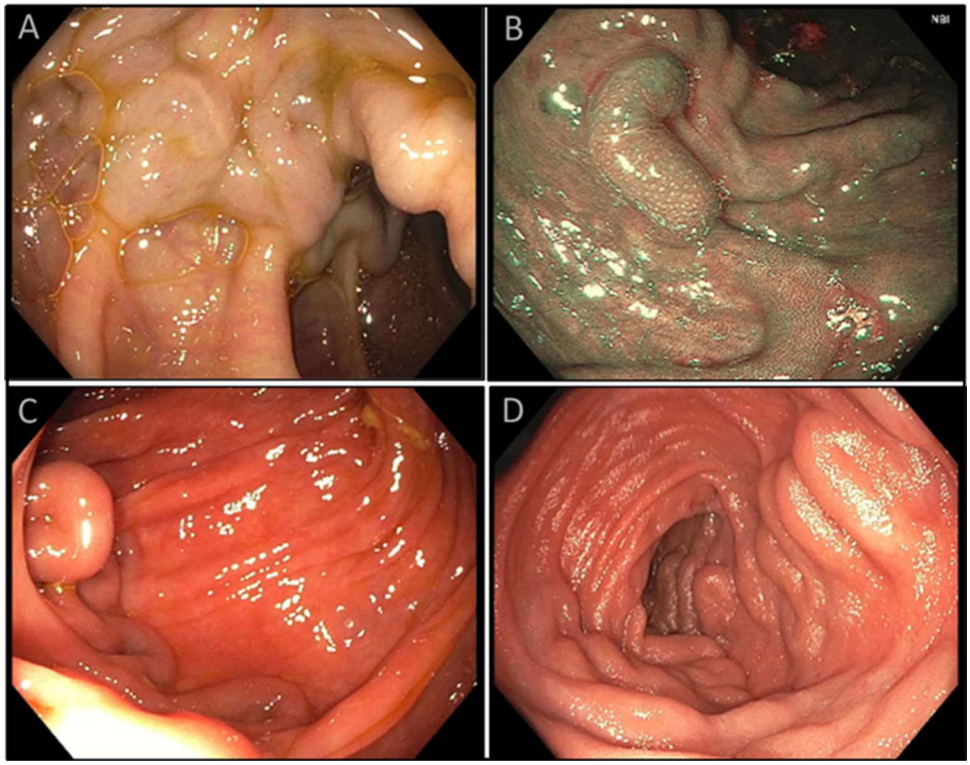

Figure 3. Colonoscopy images of colonic and ileal varices in a 38-year-old woman. (A) Rectum, (B) ascending colon (narrow band imaging), (C) transverse colon, and (D) terminal ileum.

years of life.[5,7,14] There is an increased incidence of familial involvement, with approximately 15–20 cases reported.[13]

The underlying pathology resulting in portal hypertension can be categorized by the level of impairment and includes prehepatic, hepatic, and posthepatic etiologies.[18] Prehepatic cases most commonly result from portal vein thrombosis or congenital vascular abnormalities. Hepatic cases include intrinsic liver pathology and are further divided into presinusoidal (granulomatous disease, hepatoportal sclerosis, and infectious disease), sinusoidal (cirrhosis, steatohepatitis, and amyloidosis), or postsinusoidal (sinusoidal obstructive syndrome) disease. Posthepatic cases result from alterations in blood flow proximal to the liver, such as Budd-Chiari syndrome and congestive heart failure. The etiology can be distinguished by examining the pressure gradient between the hepatic vein and sinusoidal wedge pressure. This gradient is often normal in prehepatic and presinusoidal disease and elevated in sinusoidal and postsinusoidal hepatic dysfunction. In this case, the mother presented with normal portal pressure gradients and normal portal vein pressures, which are most consistent with prehepatic or presinusoidal disease, specifically related to congenital vascular anomalies.[19]

Data are limited regarding patient management and long-term outcomes for idiopathic varices because of low prevalence. Most cases are managed conservatively, and active treatment is considered in the presence of recurrent or significant gastrointestinal bleeding. Successful treatments in emergent cases have included partial colectomy, venous coil embolization, balloon-occluded retrograde transvenous obliteration, or angioplasty in cases with vascular involvement.[12,16,20] Variceal banding, clipping, and sclerotherapy have also been used when varices are limited to the rectum, but they are generally avoided in proximal disease with risk of perforation.[6,15,21] Alternatively, beta-blockers can be used to reduce portal pressures, similar to esophageal varices.[17,20]

DISCLOSURES

Author contributions: All authors contributed to manuscript drafting and editing; all authors have approved the final version of the manuscript. J. Gallagher is the article guarantor.

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