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Short Report

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"My belly hurts and it's hard to breath": A case report of Osler-Weber Rendu syndrome

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Background

Osler-Weber-Rendu Syndrome (OWRS) also known as Hereditary Hemorrhagic Telangiectasia (HHT) is an autosomal dominant vascular disorder that causes epistaxis, Arteriovenous Malformations (AVMs), and telangiectasias. While Gastrointestinal (GI) bleeding is a known and relatively common effect of OWRS, less common findings such as ischemic colitis have been reported. Here, we present a case of a patient with underlying OWRS who was found to have ischemic colitis.

Case presentation

Patient was a 64-year-old male with underlying OWRS and one previous episode of bleeding AVMs in his lower GI tract requiring cauterization two years prior, who presented with right upper quadrant abdominal pain, diarrhea, and worsening dyspnea of one-week duration. Contrast-enhanced CT abdomen demonstrated colitis, with thickening of the hepatic flexure of the colon and mesenteric stranding. The patient was admitted for further workup of colitis and progressive dyspnea.

Initial workup was not convincing for an infectious etiology of the colitis, with no leukocytosis or lactic acidosis, and unremarkable CMP. Moreover, despite a normal lactic acid on admission, the patient had abdominal pain out of proportion to his physical exam, a low hemoglobin of 7.4 g/dL and a positive fecal occult blood test. He was transfused two units of Packed Red Blood Cells (pRBCs) and his hemoglobin stabilized at 9 g/dL. The patient then underwent colonoscopy, which revealed segmental moderate inflammation in the ascending colon characterized by congestion, erosions, erythema, friability and shallow ulcerations in the ascending colon consistent with ischemic colitis. Biopsies of the ulcers demonstrated acute inflammation without signs of inflammatory bowel disease.

The patient was also clinically fluid-overloaded at admission, with a chest radiograph showing bilateral pulmonary congestion. NT-proBNP was elevated at 1498 pg/mL. A transthoracic echocardiogram revealed moderate pulmonary hypertension (RVSP 64.2 mmHg) and an estimated ejection fraction of 60-65% (cardiac index 3.96 L/min/m^2).

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Treatment

The patient was managed with IV ciprofloxacin and metronidazole, judicious IV fluid resuscitation followed by gentle diuresis, bowel rest, and received a total of two units of pRBCs. He was hospitalized for four days and by the date of discharge had complete resolution of his dyspnea and abdominal pain.

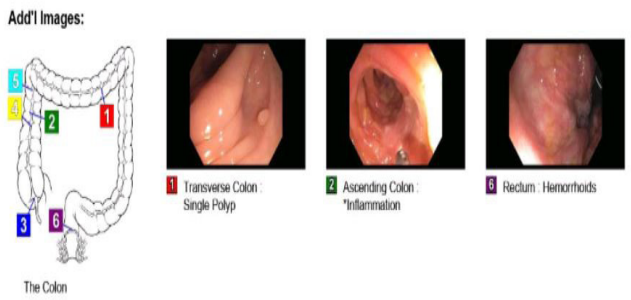

Figure 1: Findings from colonoscopy displaying the transverse colon, ascending colon showing inflammatioin, and rectal hemorrhoids.

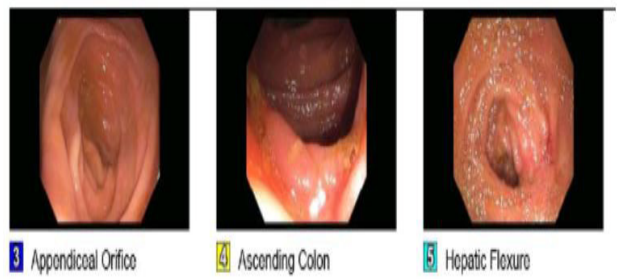

Figure 2: Findings from colonoscopy showing the appendiceal orifice, ascending colon, and hepatic flexure.

Discussion

OWRS is an autosomal dominant disorder with symptoms ranging from asymptomatic, to recurrent epistaxis, AVMs, GI bleeding and complications of high-output heart failure, iron deficiency anemia and portal hypertension. This case is an example of a patient with known OWRS and previous AVMs developing an acute GI bleed requiring blood transfusion, with simultaneous high-output heart failure and ischemic colitis. This case demonstrates the importance of considering more than just GI bleed in the differential when a patient with OWRS presents with abdominal pain and positive fecal occult. A differential diagnosis such as ischemic colitis must be considered as an alternative or concurrent diagnosis. Finally, the chronic anemia resulting from AVMs places patients at risk for high-output heart failure resulting in dyspnea. Awareness of ischemic colitis as a cause of abdominal pain in patients with OWRS is important and prompt evaluation with CT imaging may be warranted despite normal lactic acid levels.

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