Klippel–Trenaunay syndrome: endoscopic findings

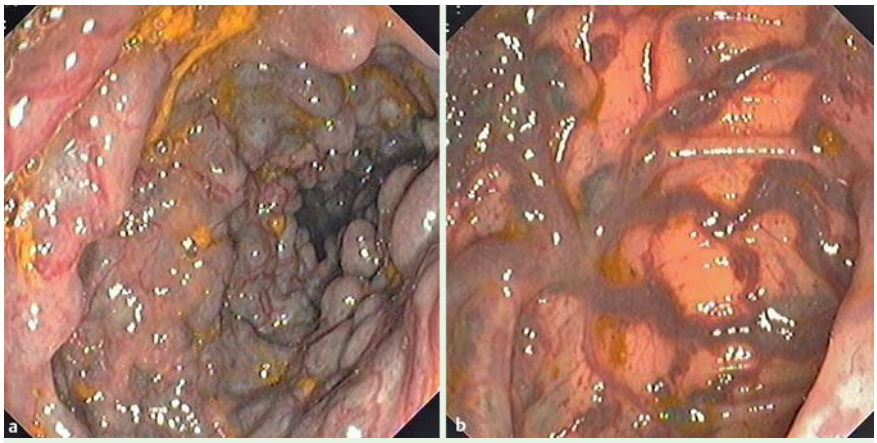


Fig. 1 **a, b** Endoscopic finding of ectasia and congestion of the submucosal and mucosal venous vessels and hemangiomas affecting the rectum and the left colon in a 44-year-old man with Klippel–Trenaunay syndrome.

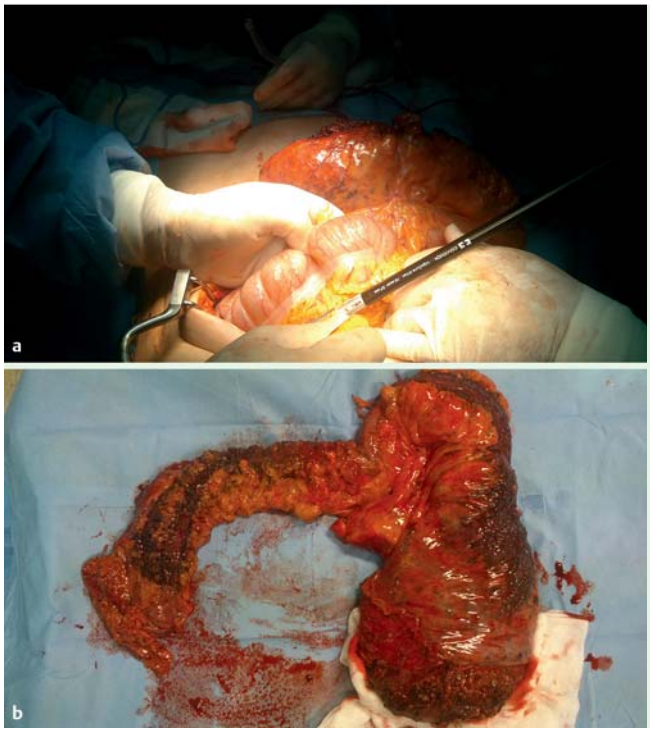
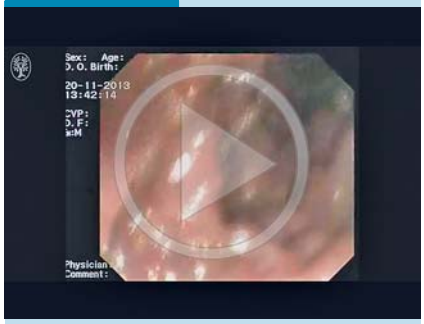


Fig. 2 Intraoperative findings of (**a**) normal vascular pattern in the transverse colon and (**b**) numerous venous varicosities in the rectum and the left colon.

Video 1



Klippel–Trenaunay syndrome: endoscopic findings in a 44-year-old man. Colonoscopy revealed ectasia and congestion of the submucosal and mucosal venous vessels and hemangiomas with a continuous pattern, affecting the rectum and the left colon with normal vessels in the transverse colon.

A 44-year-old man with no relevant past medical history was referred for colonoscopy because of recurrent hematochezia and iron deficiency anemia. He reported no abdominal pain or weight loss. He had been on oral therapy with ferrous sulfate. A hypertrophic lower left limb was seen on physical examination. Colonoscopy (**Video 1**) revealed ectasia and congestion of the submucosal and mucosal vessels and hemangiomas with a continuous pattern, affecting the rectum and the left colon, with normal vessels in the transverse colon (**Fig. 1**). These vascular lesions were not bleeding actively at the time of endoscopy, but were the likely cause of the gastrointestinal bleeding.

Esophagogastroduodenoscopy did not reveal any bleeding lesions. Abdominal contrast-enhanced tomography showed concentric thickening of the rectum and the left colon, due to multiple venous ectasias, and anomalous vascular structures in the right gluteal region. A presumptive diagnosis of Klippel–Trenaunay syndrome was made, based on the presence of soft tissue venous malformations (gluteus) associated with colonic venous malformations and tissue hypertrophy. Endoscopic treatment was not feasible because of the extensive and circumferential nature of the colonic vascular lesions. Angiography with selective venous embolization was proposed, but was not performed because of technical limitations. However, because of daily bleeding episodes and worsening of anemia that required multiple blood transfusions, a left hemicolectomy was performed (**Fig. 2**). The patient had no complications during the postoperative period and no recurrence of bleeding after 18 months of follow-up.

Klippel–Trenaunay syndrome is a congenital malformation syndrome characterized by the presence of cutaneous capillary malformations, asymmetrical disturbed growth of soft tissues and/or bone, and venous and/or lymphatic malformations [1]. It is rare, with an unknown etiology and an incidence of approximately 1:100 000 live births [2]. Gastrointestinal involvement occurs in about 1%–13% of affected patients, most of whom are asymptomatic [3]. Gastrointestinal hemorrhage can occur, ranging from occult to life-threatening and severe in nature [2, 3]. In this patient, endoscopic and angiographic treatments were not feasible and the severity of symptoms forced an early surgical approach. The endoscopic findings associated with Klippel–Trenaunay syndrome are highlighted.

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Competing interests: None

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Bibliography
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