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Severe anal bleeding in Proteus syndrome: a case report

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Abstract Proteus syndrome was originally described by Cohen and Hayden in 1979. The disorder was named Proteus syndrome by Wiedmann and colleagues in 1983 after Proteus, the giant Greek god of the sea. Proteus syndrome is a rare, sporadic, congenital polymorphic condition. Approximately 200 cases have been reported in the literature, but none has been associated with anal bleeding from hemorrhoids. We describe the case of a 21-year-old man with Proteus syndrome with severe anal bleeding. A hemorrhoidectomy was assumed to be too risky because of the massive venous abnormalities seen on CT. The patient was successfully treated by Doppler-guided haemorrhoidal artery ligation (DG-HAL). Six months after surgery, the patient has had no further episodes of anal bleeding.

Key words Proteus • Hemorrhoids • DG-HAL • Anal bleeding • Venous anomalies

Introduction

Proteus syndrome was originally described by Cohen and Hayden [1] in 1979. The disorder was named “Proteus syndrome” by Wiedmann and colleagues [2] in 1983 after Proteus, the giant Greek god of the sea. Proteus could predict the future but did not always want to reveal information. Luckily he was able to change the shape of his body to avoid capture. The name Proteus therefore refers to polymorphism.

Proteus syndrome is a rare congenital condition [3]. Somatic mosaicism, lethal in the nonmosaic state, is the best working hypothesis. Although Proteus syndrome data are consistent with this hypothesis, it has not been proven. Its etiology is unknown to date [4]. The clinical manifestations include partial gigantism of the hands or feet, hemihypertrophy, macrodactyly, plantar or palmar hyperplasia, hemangioma, lipoma, lymphangioma, varicosity, epidermal and connective tissue nevi, cranial exostosis, macrocephaly and skeletal anomalies [5, 6]. It is now thought that the “Elephant Man” suffered from Proteus syndrome instead of neurofibromatosis type 1 [7]. Although approximately 200 cases of Proteus syndrome have been reported in the literature, none has been associated with severe anal bleeding from hemorrhoids. We report such a case in a 21-year-old man.

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Case report

A 21-year-old man with Proteus syndrome complained of severe anal bleeding after defecation. The patient had been admitted several times for anemia due to anal bleeding, for which blood transfusion was necessary. Proctoscopy showed stage 3 hemorrhoids but no active bleeding. He was treated several times with Barron liga-

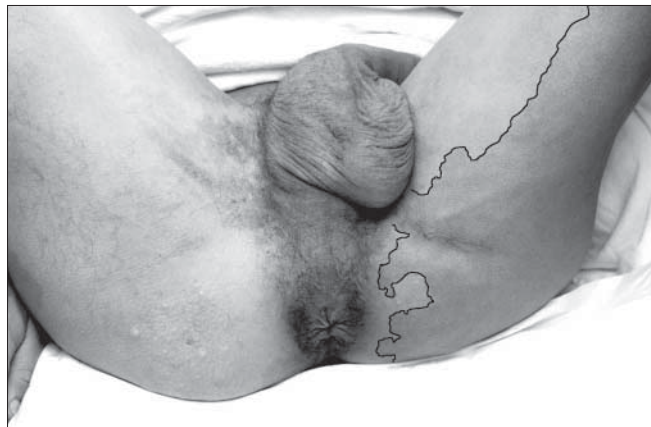


Fig. 1 A 21-year-old man with Proteus syndrome. Shown are a large portwine stain on the left leg and right-sided hemihypertrophy

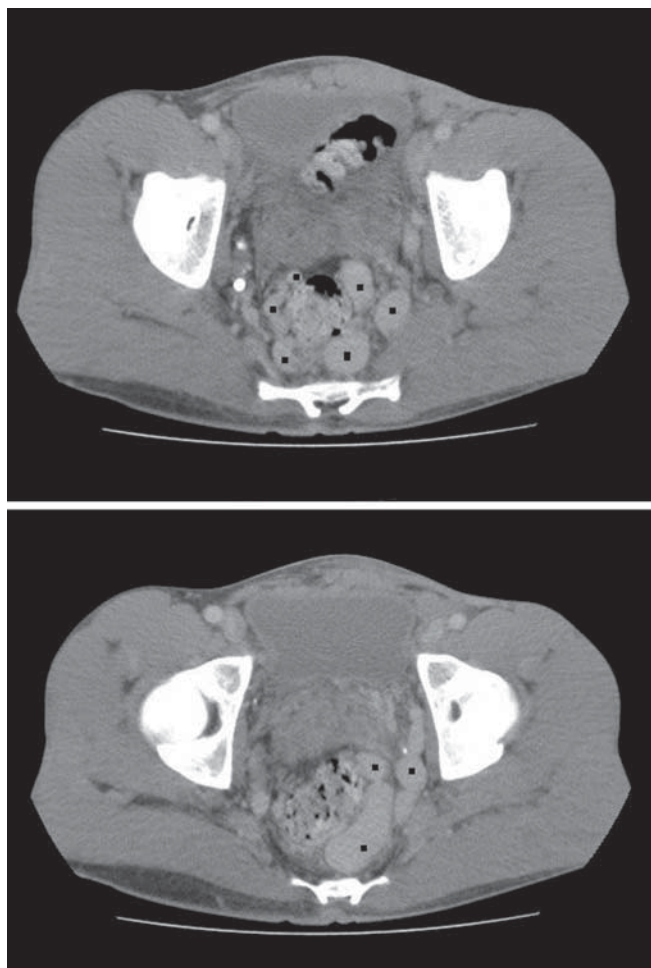


Fig. 2 CT image of the pelvis in a patient with Proteus syndrome. The dots mark the venous anomalies (dilatated veins) around the ano-rectum

tion, but this had only temporarily effects. Further symptoms of Proteus syndrome included: partial gigantism of the hands and feet, hemihypertrophy (Fig. 1), macro-

dactyly, hemangioma, lipomas, varicosis, epidermal and connective tissue nevi, and scoliosis.

Because of the repeated bleeding, computed tomography (CT) of the lower abdomen was performed, and showed massive venous anomalies around the rectum (Fig. 2). Because of the massive venous abnormalities, resection was considered to be a major risk. We assumed that the bleeding was from hemorrhoids and not from the major veins around the rectum, but that a connection between the hemorrhoids and the veins was likely.

We performed Doppler-guided hemorrhoidal artery ligation (DG-HAL), a procedure first described by Morinaga et al. [8] in 1995. With this treatment, the intraluminal arteries located 2 cm proximal to the dentate line are ligated with a special proctoscope, which contains an integrated Doppler transducer and a lateral ligation window. The internal pressure of the plexus hemorrhoidalis is decreased, because the arteries responsible for the blood inflow are ligated, and the typical symptoms of hemorrhoids disappear.

At a recent follow-up, 6 months after surgery, the anal bleeding had completely resolved.

Discussion

Vascular abnormalities as part of the Proteus syndrome have been described several times in the literature. Hoeger et al. [9] presented data on 22 patients with vascular abnormalities diagnosed on clinical grounds and supported by imaging studies or histology. Thirty-five vascular anomalies were identified in 22 patients, and more than one type of vascular anomaly was present in 10 patients. Other studies revealed vascular anomalies in 70% of the patients with Proteus syndrome. The percentage of 70% is probably higher due to under-reporting. They also reported that varicosities and prominent veins were the most common findings. Nguyen et al. [10] studied 24 patients in a prospective cohort study. Vascular anomalies were identified in 20 patients (83%). The vascular anomalies were mostly located in the trunk and legs. In contrast to our patient, none of the reviewed cases had anal bleeding. Our patient had severe anal bleeding from hemorrhoids, probably associated with vascular abnormalities of Proteus syndrome. He was successfully treated with Doppler-guided hemorrhoid artery ligation (DG-HAL), because a conventional hemorrhoidectomy was considered to be too risky.

In conclusion, proteus syndrome can be recognized by its specific characteristics. Severe anal bleeding in these patients can be associated with massive venous abnormalities. CT may help identify these patients. Surgical hemorrhoidectomy can be risky and should be avoided. DG-HAL can be a good alternative and is a safe and effective method to treat hemorrhoids.

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