CASE REPORT

Involvement of Splenic Hemangioma and Rectal Varices in a Patient with Klippel-Trenaunay Syndrome

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Klippel-Trenaunay syndrome (KTS) is characterized by a cutaneous vascular nevus of the involved extremity, bone and soft tissue hypertrophy of the extremity and venous malformations. We present a case of KTS with splenic hemangiomas and rectal varices. A 29-year-old woman was referred for intermittent hematochezia for several years. She had history with a number of operations for cutaneous and soft tissue hemangiomas since the age of one year old and for increased circumference of her left thigh during the last few months. Abdominal CT revealed multiple hemangiomas in the spleen, fusiform aneurysmal dilatation of the deep veins and soft tissue hemangiomas. There was no evidence of hepatosplenomegaly or liver cirrhosis. Colonoscopy revealed hemangiomatous involvement in the rectum. There were rectal varices without evidence of active bleeding. Upon venography of the left leg, we also found infiltrative dilated superficial veins in the subcutaneous tissue and aneurysmal dilatation of the deep veins. The patient was finally diagnosed with KTS, and treated with oral iron supplementation only, which has been tolerable to date. Intervention or surgery is not required. When gastrointestinal varices or hemangiomatous mucosal changes are detected in a young patient without definite underlying cause, KTS should be considered. (Korean J Gastroenterol 2011;58:157-161)

Key Words: Klippel-Trenaunay-Weber syndrome; Hemangioma; Varicose veins

INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is a rare but relatively well-documented congenital malformation characterized by the triad of capillary and venous malformations, in addition to bony or soft tissue hypertrophy of the affected limb. The clinical features of KTS are variable and have been reported worldwide. Vascular malformations involving the gastrointestinal tract have also been reported rarely and can be a source of significant morbidity and even mortality. Thus, different approaches to manage the bleeding sources are needed. KTS with splenic hemangiomas has not been reported in Korea yet. We report here a case of KTS in a 29-year-old woman who had rectal varices and multiple splenic hemangiomas. A literature review on KTS is also presented.

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CASE REPORT

A 29-year-old woman was referred to our institution with recurrent hematochezia. She had suffered from intermittent hematochezia for several years. She had a gait disturbance and operative scars on the left shin. She had had a number of operations for hemangioma of the abdomen at age 1, the left ankle at age 7 and the left lower leg in adolescence. She had used lightweight orthosis on the left ankle because of the development of foot drop 7 years ago. The circumference of the left thigh had increased during the last several months.

She felt more comfortable flexing her left leg than standing up and slapping her foot down onto the floor. We could not find any signs of family history of hepatitis, liver cirrhosis, limb deformity or hemangioma.

On physical examination, the liver and the spleen were not palpable and the abdomen was flat and soft. At the time of admission, basic metabolic analyses, liver function tests, and complete blood count analyses were performed. All parameters were within normal limits with the exception of Hb 10.4 g/dL (MCV 80.7fL, MCH 23.9 pg), Fe 15 μg/dL, and ferritin 3.8 ng/mL. An abdominal CT scan showed multiple hemangiomas in the spleen and fusiform aneurysmal dilatation of the left common and internal iliac veins without proximal obstructive lesions (Fig. 1). Additionally, there were diffuse mural thickenings with multifocal calcifications involving rectum compatible with rectal varices and phleboliths, and multifocal delayed enhancing nodular lesions in the left buttoc and gluteus muscle with several calcifications suggesting soft tissue hemangioma with phleboliths. There was no evidence of hepatosplenomegaly, liver cirrhosis or ascites. The outcome of an esophagogastroduodenoscopic investigation was unremarkable. However, colonoscopy showed the loss of the normal submucosal vascular pattern. There were hemangiomatous involvement and variceal change in the rectum without evidence of active bleeding (Fig. 2). The blood vessels in the lower sigmoid colon were also dilated. Venography of the left leg showed dilated superficial veins in the subcutaneous tissues and aneurysmal dilatation of the

Fig. 1. An abdominal CT scan revealed multiple hemangiomas in the spleen (A), and fusiform aneurysmal dilatation of the left common (B) and internal iliac veins (C).

Fig. 2. Colonoscopy showed hemangiomatous involvement in the rectum; there were rectal varices, but no evidence of active bleeding.
left common and internal iliac veins (Fig. 3).

Given the degree of anemia, the age of the patient, and her current quality of life, she has been treated with oral iron supplementation only. No additional interventions or additional surgery have so far been necessary.

**DISCUSSION**

KTS was originally described by the French physicians Klippel and Trenaunay in 1900. While it is characterized by a cutaneous vascular nevus of the involved extremity (capillary malformation), bone and soft tissue hypertrophy of the extremity and venous malformations or varicose veins, KTS can be diagnosed on the basis of any two of these features.1

Although KTS is a relatively well-documented disease, with some large-scale studies reported,1,3 there are few case reports in Korea. Recently, rectal involvement, esophageal and gastric varices, and sigmoid varices were reported in Korea.4 The present case involves a patient who presented with multiple cutaneous hemangioma, soft tissue hemangioma and hypertrophy, rectal varix, splenic hemangioma, and deep venous aneurysmal dilatation, all of which are compatible with a diagnosis of KTS. Nevertheless, the fact that some of these clinical features had existed since her childhood, and that she was barely diagnosed with KTS at the age of 29, suggests the point that this syndrome could be underdiagnosed.

Gastrointestinal involvement occurs in 1% to 12.5% of KTS patients.2 The most common involvement in the gastrointestinal tract is diffuse cavernous hemangiomas of the distal colon and the rectum.5,6 In one of the largest published studies of KTS patients, hematochezia was observed in six of the 588 patients.3 Although rare, bleeding from jejunal hemangiomas and esophageal and gastric varices also have been reported.2,4,7 The clinical spectrum of the gastrointestinal bleeding may vary from asymptomatic occult bleeding to life-threatening massive bleeding. Gastrointestinal hemorrhage usually presents in the first decade of the patient’s life and tends to be intermittent.8 In the present case, the patient had suffered from recurrent hematochezia for several years and was diagnosed with rectal varix and iron deficiency anemia. In general, hematochezia may be caused by the posterior or compensatory venous pathways of the extremity, which drain into the internal vein.2,8 If these veins can no longer drain efficiently into the dilated internal iliac vein, rectal bleeding may occur. Increased venous pressure on defecation or mucosal trauma resulting from the passage of feces can worsen the recurrent hematochezia.9

Treatment plans for colorectal hemangiomas in KTS should be individualized on the basis of the extent and severity of blood loss. Supportive management and iron supplements can be sufficient for those patients who only experience the occasional insignificant bleeding. The need for transfusion after massive bleeding or worsening of the quality of life can suggest surgical treatment or an intervention to the involved bowel segment is required. Conservative managements such as endoscopic therapy with sclerosis, band ligation, and laser are usually ineffective due to the extensive nature of the lesion.10 Thus, endoscopic therapeutic modalities are generally used for postoperative residual sites or lo-
calized lesions.\textsuperscript{11} The neodymium: yttrium-aluminum-garnet laser for residual lesions and argon laser photoagulation for hemangiomas involving the distal 7 cm of the anorectum were effective. Our patient was treated with oral iron supplement only because of no evidence of active bleeding.

In addition to the gastrointestinal tract, other visceral organ involvements such as the liver, bladder, spleen, kidney, heart and lung have also been reported.\textsuperscript{3} CT of the abdomen and pelvis is a simple, noninvasive method by which visceral hemangiomas can be found.\textsuperscript{12}

Phleboliths are also known as pathognomonic features of venous malformations in very young patients and may be manifestations of previous hemorrhage or thrombus.\textsuperscript{13}

The involvement of the spleen is rare and can be due to the presence of hemangioma or lymphangioma.\textsuperscript{14} Percutaneous biopsy of spleen is not advisable because of the high risk of hemorrhage. Noninvasive imaging with ultrasonography, MRI, or CT, and lymphoscintigraphy is used for diagnosis of these lesions. Complications of splenic hemangiomas include hypersplenism, rupture and malignant degeneration.\textsuperscript{11} Fortunately, since recent reviews of adult patients have revealed that asymptomatic patients with a small splenic hemangioma (\(<\ 4\ \text{cm})\) can be treated conservatively with observation,\textsuperscript{15} we also decided to follow this approach with our patient. KTS with splenic hemangiomas has not been reported in Korea yet.

Our patient had deep venous aneurysmal malformations. This abnormality can lead to thrombotic episodes in the extremities and multiple recurrent pulmonary emboli.\textsuperscript{16} One study reported that 2.2\% of the KTS patients who were enrolled in the study experienced a venous thrombo-embolism.\textsuperscript{17} In these conditions, oral contraceptives should be strongly avoided. The physician may have to warn their patients that pregnancy can aggravate the symptoms, resulting in either lower extremity swelling, venous malformations, varicosities or venous insufficiency.\textsuperscript{1} In addition, a popliteal venous aneurysm can induce foot drop because of common peroneal nerve compression,\textsuperscript{18} although we failed to detect such an aneurysm in our patient. Since the foot drop can be reversed by proper surgical intervention, when a patient with KTS complains of being unable to turn the ankle and toes upward, the physician has to consider performing venography to rule out the possibility of a popliteal venous aneurysm.

While many suggestions regarding the underlying cause of KTS have been proposed, the pathogenesis of KTS remains obscure.\textsuperscript{1} However, it is most likely to be a generalized mesodermal development abnormality.\textsuperscript{19} In addition, several genetic mutations including the recently reported E133K mutation, may be at least partly responsible for the heterogeneous mesodermal defect.\textsuperscript{20}

We report here a case of a patient with Klippel-Trenaunay syndrome who had rectal varix and multiple splenic hemangiomas, yet had not been diagnosed until the age of 29 years. While this patient has had a benign clinical course to date, the disease can sometimes show fatal gastrointestinal complications such as massive hemorrhage. Therefore, when endoscopists encounter gastrointestinal varix or hemangiomatous mucosal change in young patients in the absence of a definite underlying cause, it is important to consider the possibility of KTS and to evaluate the patient further by at least performing CT and carefully obtaining the medical history of the patient.

REFERENCES