Case Report
pISSN 1738-2637
J Korean Soc Radiol 2012;67(4):289-292

대한영상의학회지

Rectal and Splenic Vascular Malformation in Klippel-Trenaunay-Weber Syndrome: A Case Report[1]

직장과 비장의 혈관기형을 동반한 Klippel-Trenaunay-Weber Syndrome: 증례 보고[1]

Ha Youn Kim, MD[1], Yun-Woo Chang, MD[1], Dong Hwan Lee, MD[2]

Departments of [1]Radiology, [2]Pediatrics, Soonchunhyang University Hospital, Seoul, Korea

Klippel-Trenaunay-Weber syndrome (KTWS) is a rare congenital disorder, characterized by a cutaneous vascular nevus of the involved extremity, vascular malformations, bone and soft tissue hypertrophy of the extremity. We present the case of an 18-year-old female patient with KTWS, showing a marked rectosigmoid wall thickening and phlebolith, and also variable sized cystic masses in the spleen, as a result of vascular malformations.

Index terms
Klippel-Trenaunay-Weber Syndrome
Vascular Malformations
Computed Tomography

Received April 9, 2012; **Accepted** July 26, 2012
Corresponding author: Yun-Woo Chang, MD
Department of Radiology, Soonchunhyang University Hospital, 59 Daesagwan-ro, Yongsan-gu, Seoul 140-743, Korea.
Tel. 82-2-709-9396 Fax. 82-2-709-3928
E-mail: ywchang@schmc.ac.kr

INTRODUCTION

Klippel-Trenaunay-Weber syndrome (KTWS) is a rare congenital malformation, characterized by cutaneous hemangioma, hypertrophy of bone and soft tissue, varicose veins, and a low flow vascular malformation (1-8). The diagnosis of KTWS can be made when any two of the three features are present. It has no racial predilection, affects males and females equally, and manifests at birth or during childhood (1). The clinical features of KTWS are variable and have been extensively documented. Vascular malformations, associated with KTWS, are located on the extremities, visceral organs, or spinal canal. Vascular malformations typically occur on the lower extremity and can occasionally be bilateral or can involve the upper extremity (1-4). Vascular malformation, involving the gastrointestinal tract, has been rarely reported with the clinical symptom of hematochezia, and can be one of the causes of significant morbidity or life threatening emergencies in KTWS (1-3). KTWS with vascular malformation, involving the spleen, has been rarely reported. We report a case of KTWS in an 18-year-old girl, who had low flow vascular malformation, involving the rectum, sigmoid colon and spleen.

CASE REPORT

An 18-year-old female patient was admitted to our institution with recurrent hematochezia. She had suffered from intermittent hematochezia and pain in the right calf for several years. On physical examination, she had a cutaneous vascular nevus on the right ankle, soft tissue hypertrophy, varicose veins on the right thigh and calf (Fig. 1A, B). The liver and spleen were not palpable and the abdomen was flat and soft.

She had a history of several operations for lymphatic malformations in the pelvic cavity and venous malformation on the right buttock and foot, since the age of two years old. She had an increased circumference of her right leg during childhood. She experienced pain in her lower extremities after walking for a few minutes. On venography examination at the age of 14

years, she had varicose veins on the right thigh and calf (Fig. 1C). On MR imaging at the age of 14 years, she showed diffuse venous and lymphatic vascular malformations in pelvic cavity and bilateral buttock (Fig. 1D).

Laboratory findings revealed: Hemoglobin 6.5 g/dL (12-16 g/dL), hematocrit 19.6% (36-48%), and platelet 267 × 10^3/μL (130-450 × 10^3/μL).

The abdominopelvic computed tomography (CT) scan showed marked mural thickening of the sigmoid colon and rectum with engorged enhancing vascular structures, and punctuate multiple calcifications, consistent with vascular malformations and phleboliths (Fig. 1E, F). Vascular malformations were also identified in mainly right subcutaneous tissue, buttock muscle and pelvic cavity, without evidence of arterio-venous fistula. There were multiple, variably sized, small low attenuation cystic masses on the spleen without contrast enhancement (Fig. 1G). There was no evidence of hepatosplenomegaly or ascites. The colonoscopy examination revealed vascular malformations, involving the rectum and sigmoid colon, and normal appearance of the ascending colon (Fig. 1H).

Taking into consideration of the mild anemia and young age, she was treated with transfusion and oral iron supplementation. At the 7 day follow-up, the hemoglobin level and clinical symptom of hematochezia improved.

DISCUSSION

Klippel and Trenaunay first described a syndrome characterized by a capillary nevus of the affected extremity, lateral limb hypertrophy, and varicose veins. Weber noted the association of these findings with arteriovenous malformation in the 1900s (1). If the patients of Klippel-Trenaunay syndrome (KTS) have clini-

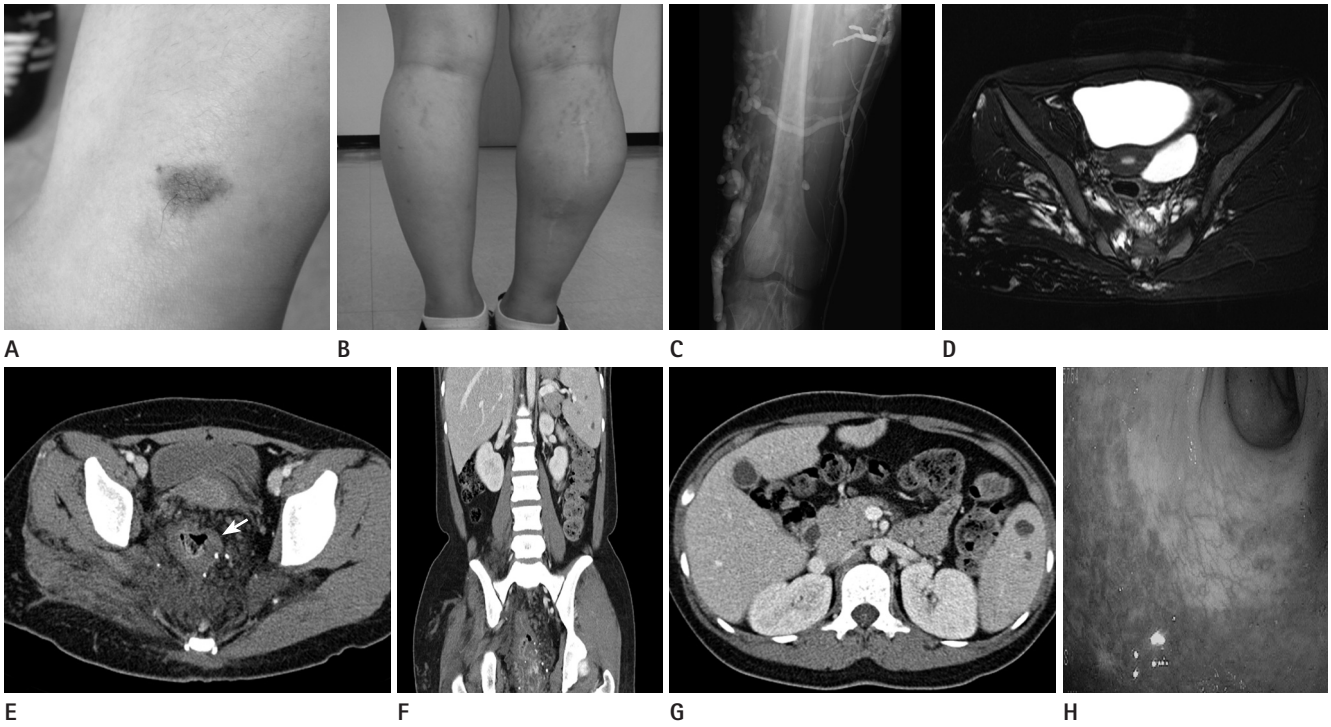

Fig. 1. An 18-year-old female patient diagnosed with Klippel-Trenaunay-Weber syndrome.
A. Cutaneous hemangioma on the right ankle.
B. Unilateral hypertrophy of soft tissue is noted in the right calf area.
C. Venography of the right leg obtained 14-year-old shows dilated superficial veins in the subcutaneous tissue.
D. Axial T2-weighted fat suppression magnetic resonance image obtained 14-year-old shows diffuse high signal intensity venous and lymphatic vascular malformations in the pelvic cavity, bilateral muscular compartment and subcutaneous tissue.
E, F. Axial **(E)** and coronal reconstruction CT **(F)** images show diffuse recto-sigmoid wall thickening (arrow), phleboliths and tubular enhanced vascular engorgement as a result of vascular malformation. Also note numerous venous malformations in the right buttock.
G. Axial CT scan shows multiple low density cystic masses in spleen without contrast enhancement, suggestive of lymphatic malformations of the spleen.
H. Colonoscopy shows vascular malformation involved the recto-sigmoid colon.

cally significant arteriovenous malformation, the patients are considered as a separate condition of KTS, called Parker-Weber syndrome. Some authors used the term KTWS to describe the conditions of KTS, who have clinically significant arteriovenous malformation (1). KTWS is defined as a combination of cutaneous hemangiomas, soft tissue or bone hypertrophy, and varicose veins or low flow vascular malformations (1-6).

The pathogenesis of KTWS remains obscure. It is most likely to be a generalized mesodermal development abnormality. Several genetic mutations have been described in the angiogenic protein, including VG5Q or the recently reported E133K mutation. However, the genetic basis of KTWS is far from being solved (1, 3).

According the International Society for the Study of Vascular Anomalies classification, which is widely accepted, vascular malformation are subdivided into slow-or low-flow and fast-or-high flow malformations. KTWS is defined as combined capillary, lymphatic, and venous, low-flow vascular malformation in an overgrown limb (7). Vascular malformation of the gastrointestinal tract occurs in 1% to 12.5% of KTWS patients (3, 4). However, it may occur in 20% of KTWS patients and may go unrecognized in patients without overt symptoms. The most common sites of gastrointestinal tract involvement in these patients are the distal colon and rectum. In these patients, clinically significant gastrointestinal bleeding to lead massive hematochezia could be the manifestation, and it could induce consumptive coagulopathy (1).

Radiologic evaluation plays an important role in the diagnosis and ongoing evaluation of KTWS. Sonography may be used to identify the abnormal varicous veins. Abdominopelvic CT scan is a noninvasive method for assessing visceral vascular malformations. Phleboliths in a very young patient are pathognomonic for venous malformations and are manifestations of prior hemorrhage or thrombus. Magnetic resonance imaging (MRI) is performed to evaluate the extent of vascular malformations in KTWS (1, 5). Clinical or radiologic findings of Parkes-Weber syndrome could be differentiated with that of predominent venous malformations with arerior-venous fistula.

In case of hemorrhage, which requires surgical intervention, preoperative angiography is required to define the anatomy and extent of intestinal involvement to guide surgical resection (1, 4). Endoscopy is essential procedure to detect the bleeding focus and also useful for determining the extent of gastrointestinal tract involvement of KTWS (2).

In rare cases, other viscera may be involved in KTWS, including the spleen, lung, heart, liver and kidney (2). Splenic involvement in KTWS is very rare and splenic hemangioma or lymphangioma have been rarely reported. Percutaneous biopsy of the spleen is not recommended because of the high risk of hemorrhage. Noninvasive radiological evaluation with sonography, CT, or MRI, and lymphoscintigraphy is used for diagnosis of the splenic lesions (3, 8). In our case, non-enhancing, multiple, variably sized low attenuation cystic masses, representing multiple lymphangiomas, were detected in the spleen without splenomegaly on CT scan.

Management of gastrointestinal vascular malformation in KTWS depends on the extent and severity of bleeding. Conservative managements and iron supplements for iron deficiency anemia may be sufficient in patients who presents with occasional non-significant bleeding. Resection of the involved bowel segment is usually necessary to adequately control the bleeding to save the patients' lives (1, 2). Although KTWS is a rare disease, causing lower gastrointestinal tract bleeding, physicians should be aware of those disease entity and proper radiologic evaluation should be done for diagnosis of the disease and decision making in patient management.

REFERENCES

1. Cha SH, Romeo MA, Neutze JA. Visceral manifestations of Klippel-Trénaunay syndrome. *Radiographics* 2005;25:1694-1697
2. Cheon SH, Lee SH, Park EB. Rectal involvement of Klippel-Trenaunay syndrome. *J Korean Soc Coloproctol* 2009;25:52-55
3. Choi YJ, Jee SR, Park KS, Ryu CH, Seo HR, Ha SI, et al. [Involvement of splenic hemangioma and rectal varices in a patient with klippel: trenaunay syndrome]. *Korean J Gastroenterol* 2011;58:157-161
4. Wilson CL, Song LM, Chua H, Ferrara M, Devine RM, Dozois RR, et al. Bleeding from cavernous angiomatosis of the rectum in Klippel-Trenaunay syndrome: report of three cases and literature review. *Am J Gastroenterol* 2001;96:2783-2788

5. Kanterman RY, Witt PD, Hsieh PS, Picus D. Klippel-Trenaunay syndrome: imaging findings and percutaneous intervention. *AJR Am J Roentgenol* 1996;167:989-995
6. Park MH, Kwon ST, Shin BS, Kim YM. An arteriovenous malformation in the suprapatellar fat pad of the knee associated with Klippel-Trenaunay-Weber syndrome: a case report. *J Korean Radiol Soc* 2006;54:27-31
7. Lowe LH, Marchant TC, Rivard DC, Scherbel AJ. Vascular malformations: classification and terminology the radiologist needs to know. *Semin Roentgenol* 2012;47:106-117
8. Yamazaki M, Kawamura Y, Ohka T, Katada S, Morita K, Nakagawa M, et al. Cavernous lymphangioma of the spleen in a patient with Klippel-Trenaunay-Weber syndrome. *Intern Med* 1994;33:574-577

직장과 비장의 혈관기형을 동반한 Klippel-Trenaunay-Weber Syndrome: 증례 보고[1]

김하연[1] · 장윤우[1] · 이동환[2]

Klippel-Trenaunay-Weber syndrome (이하 KTWS)은 드문 선천성 질환으로 피부의 혈관종, 사지비대, 정맥류나 혈관기형을 특징으로 한다. 저자들은 18세 여아에서 S결장과 직장 벽의 비후와 정맥석 및 비장에서 다양한 크기의 낭종으로 보이는 혈관기형을 가지는 KTWS 증례를 보고하고자 한다.

순천향대학교병원 [1]영상의학과, [2]소아과