SIR 2005 Annual Meeting Film Panel Case: Klippel-Trénaunay-Weber Syndrome

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Abbreviations: AVM = arteriovenous malformation, KTS = Klippel-Trénaunay syndrome, KTWS = Klippel-Trénaunay-Weber syndrome

HISTORY

A 49-year-old man was referred to the interventional radiology clinic because of severe right lower-extremity varicosities. He brought to his appointment an arteriogram (**Fig 1**) and a descending venogram (**Fig 2**) obtained when he was 9 years of age. Those studies were done because of leg length discrepancy (right side longer than left). Subsequent to that, his condition was managed by keeping a prosthetic lift in his left shoe. He teaches high school woodshop and is physically active but has had increasing problems with right lower-extremity pain, heaviness, and swelling. Recently, he developed intermittent "weeping" of clear fluid between his toes and occasional right calf cellulitis, which responds to oral antibiotics. Except for wearing a support stocking, the patient has had no intervention for his vascular condition. Family history was negative.

On physical examination (**Fig 3**), the right lower extremity was larger than the left, longer and greater in girth. Large varicosities were noted in the foot, calf, and lower thigh. Signs of chronic venous insufficiency were noted in the foot and lower calf including skin thickening, discoloration, and scaliness (venous eczema; Clinical, Etiology, Anatomic, Pathophysiology classification of 4). There was no active cellulitis and no ulceration. No cutaneous vascular malformations such as "port wine" stain were noted. Lower-extremity pulses were intact. Heart sounds were normal and lungs were clear.

Subsequent to the initial visit, the patient underwent right lower-extremity venous duplex ultrasonography (US) and right lower-extremity arteriography (**Fig 4**) with ascending (**Fig 5**) and descending venography (not shown).

RADIOGRAPHIC FINDINGS

The right lower-extremity arteriogram obtained in the early 1960s was done via retrograde injection of a right femoral artery cannula. The right femoral head is larger than the left. Images demonstrate enlargement of the right iliac and femoral arteries as a result of a high-flow arteriovenous malformation (AVM) centered at the right knee and fed by multiple geniculate branches of the popliteal artery. There is early and dense opacification of the popliteal vein.

The bilateral descending venogram obtained in the 1960s was performed with simultaneous injection of femoral vein cannulas. The right leg is larger than the left. On the left, contrast material flows centrally from the injection site, opacifying the iliac veins and vena cava. On the right, there is marked reflux of contrast material into the greater saphenous system all the way to the distal calf. The saphenous system is completely without valves. There is no reflux into the deep venous system on the right, which suggests that the valvular function in the deep system was intact at that time.

The current right lower-extremity duplex US image demonstrated severe superficial and deep venous reflux of the right lower extremity extending from the groin to the ankle. There was no venous thrombosis. Varicosities in the thigh and calf communicated with the greater saphenous vein.

The current right lower-extremity arteriogram demonstrates the right knee AVM with multiple geniculate feeding branches. The vascularity of the AVM appears more dense, and the feeding branches appear larger and more tortuous than was the case 40 years earlier. The overall territory of the AVM had grown slightly over the intervening time; no new AVM was demonstrated. On the current study, there was no evidence of fracture or focal osteoporosis in the region of the AVM.

Ascending venography demonstrated that the deep venous system was intact but without valves. Descending venography demonstrated reflux into the deep and superficial venous systems to the level of the ankle.

DIAGNOSIS

Klippel-Trénaunay-Weber syndrome (KTWS); also known as Parkes-Weber syndrome.

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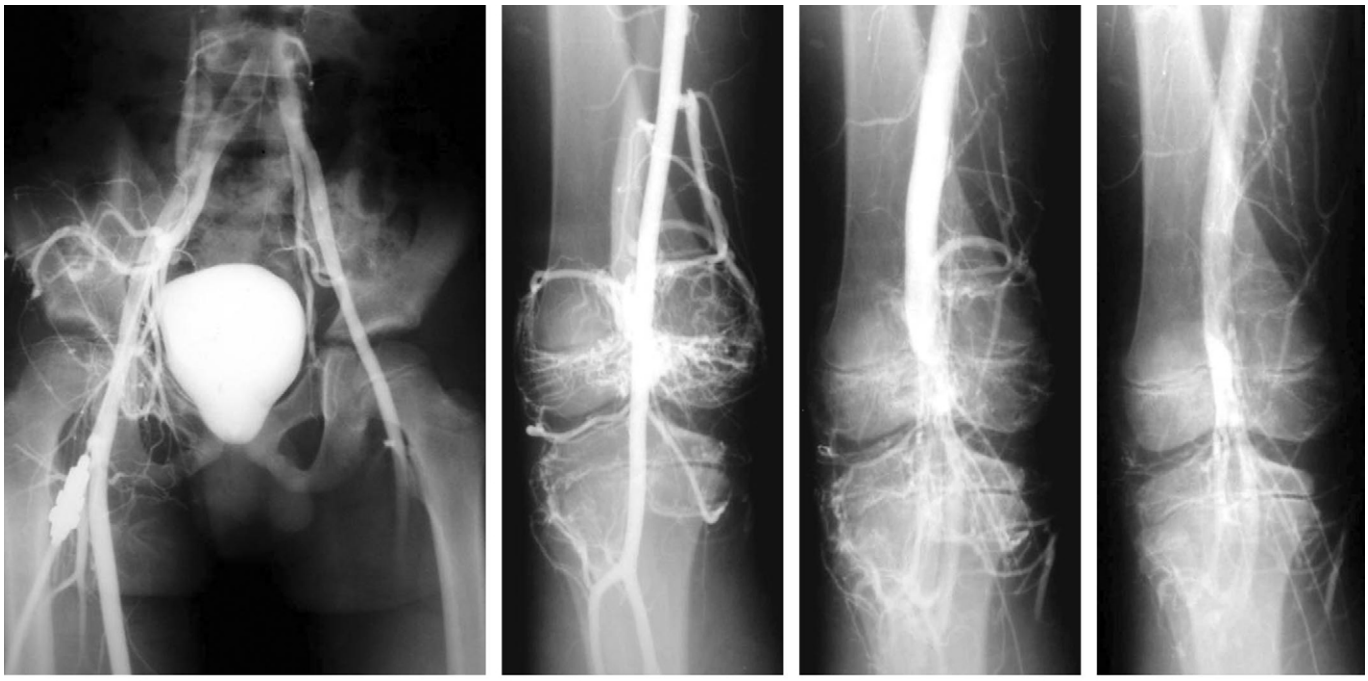

a. b. c. d.

Figure 1. Four selected images from a right lower extremity arteriogram obtained when the patient was 9 years old. Findings include hypertrophy of the right iliac and femoral arteries **(a)**, an AVM centered at the right distal femoral epiphyseal plate **(b)**, dense shunting into the popliteal vein **(b,c)**, and delayed filling of the saphenous system **(d).**

DISCUSSION

Klippel-Trénaunay-Weber Syndrome, also known as Parkes-Weber syndrome, is closely related to the less rare Klippel-Trénaunay syndrome (KTS). The names of the syndromes can be confusing. KTS was first reported in 1900 and was initially described as consisting of a triad of varicose veins, soft tissue and bony hypertrophy, and cutaneous hemangioma (1,2). In 1918, Parkes-Weber described three patients with arteriovenous fistulas in association with the Klippel-Trénaunay triad (2,3). Some authors use the name "Parkes-Weber" to describe this syndrome whereas others prefer the name "Klippel-Trénaunay-Weber." I prefer the latter because it cues the reader to all components of the condition.

The names are not all that is confusing about KTS and KTWS. Our understanding of the underlying pathology has evolved in the past 100 years, as has the medical terminology used to describe it. In addition, clinical manifestations of the syndromes can vary enormously among individual patients.

Current understanding is that KTS consists of three cardinal elements: (i) developmental venous abnormalities that include anatomic variants, varicosities, and/or malformations; (ii) soft tissue and bony hypertrophy; and (iii) capillary malformations typically manifested as cutaneous port wine stains (4–6). KTS usually affects one or more limbs (most commonly a single lower extremity) but can affect a portion of the trunk or the head. In the trunk, the abnormality does not usually cross the midline. The gastrointestinal tract or genitourinary tract may be affected (7,8). Lymphatic abnormalities may also occur (9).

KTS is a congenital developmental abnormality but is not inherited. It does appear to be associated with an angiogenic genetic mutation (4,10). Diagnosis is made clinically and must include at least two of the triad's three components. Approximately two thirds of patients have all three clinical features whereas one third have two features (4,5). Cutaneous capillary malformation is the most common manifestation, occurring in nearly 100% of cases, whereas venous abnormalities and hypertrophy each occur in approximately 60%–70% of cases (5).

A wide range of venous abnormalities can occur with KTS. Deep venous abnormalities include aneurysmal dilation, duplication, hypoplasia/aplasia of all or part of the deep venous system, external compression by fibrous bands, and persistent embryonic veins (5,6). Abnormalities of the superficial system include varicosities, anomalous veins, and venous malformations (5,6). It is uncertain if the venous dilation seen in the superficial and deep systems is caused by an intrinsic weakness in the vessel wall, valvular incompetence, or a combination of the two. Valvular aplasia has been described, usually in persistent embryonic veins (5).

Current understanding of KTWS is that it consists of high-flow AVMs occurring in a person with at least two components of the KTS triad (4,9). This syndrome is extremely rare and has more serious health consequences than KTS, including bleeding, ulceration, and high-output congestive heart failure resulting from the high-flow vascular malformations (11,12).