

HEMANGIOMATOSIS OF THE SPLEEN IN A PATIENT WITH KLIPPEL-TRÉNAUNAY SYNDROME

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Klippel-Trénaunay syndrome is a rare disorder characterized by a triad of port-wine stains, varicose veins, and bony and soft tissue hypertrophy usually involving an extremity. Visceral involvement in Klippel-Trénaunay syndrome is rare, but has been described in the colon, small bowel, bladder, kidney, spleen, liver, mediastinum and brain. In this paper we present the case of a 45-year-old woman with Klippel-Trénaunay syndrome in whom routine physical examination unexpectedly revealed the presence of a left upper quadrant mass. Abdominal US, contrast enhanced CT and whole-body PET-CT demonstrated multiple large cystic lesions within a massively enlarged spleen. Based on the clinical history and imaging findings diffuse hemangiomas of the spleen was suspected. This diagnosis was confirmed by pathologic examination after splenectomy.

Key-words: Spleen, diseases – Angioma.

Hemangioma is the most common primary benign neoplasm of spleen and usually represents a small localized tumor. Diffuse hemangiomas of the spleen is a variant of hemangioma in which the splenic parenchyma is widely replaced by vascular proliferations. This condition may be confined to the spleen or may occur as a manifestation of systemic angiomas. Associations of splenic hemangiomas with Beckwith-Wiedeman syndrome, Klippel-Trénaunay syndrome and blue rubber bleb naevus syndrome have been reported. In this paper we present the case of a patient with Klippel-Trénaunay syndrome with an incidentally discovered splenomegaly caused by diffuse hemangiomas. This case-report emphasizes the importance of the presence of visceral vascular malformations in patients with Klippel-Trénaunay syndrome.

Case presentation

A 45-year-old woman with Klippel-Trénaunay syndrome presented at our hospital with a 2,5 year history of daily attacks of breathlessness and palpitations, often accompanied by faintness and paresthesias in the hands and mouth. The symptom pattern was very suggestive of chronic hyperventilation attacks and further questioning revealed that the patient had been living under a lot of stress the last couple of years. The patient also mentioned a chronic left hypochondrial pain.

On physical examination chest and heart sounds were normal. Varicose veins and cutaneous capillary malformations were observed on the upper left leg along with soft tissue hypertrophy leading to an increase in the girth of the leg. Physical examination of the abdomen unexpectedly revealed the presence of a left upper quadrant mass extending from the costal margin to the pelvis which was tender to palpitation.

On abdominal ultrasound (US) multiple large cystic lesions were observed within a massively enlarged spleen (Fig. 1). Complementary contrast enhanced abdominal computed tomography (CT) demonstrated a significantly enlarged spleen with multiple large and several smaller cystic lesions involving the entire spleen (Fig. 2). A whole-body PET-CT was performed to exclude the possibility of cystic splenic metastases from an unknown primary. No FDG-captation was observed in or surrounding any of the lesions on PET-CT and no other lesions suggestive of neoplasm could be observed (Fig. 3). Routine serum biochemistry was normal without evidence of hypersplenism on the peripheral blood picture. Echinococcus serology was negative.

For diagnostic reasons, but also in view of the size of the spleen and the presence of chronic left hypochondrial pain, elective splenectomy was performed. Macroscopic and microscopic examination revealed the presence of multiple cavernous

hemangiomas with large, central fluid containing cavities, replacing the entire splenic parenchyma (Fig. 4). The patient's postoperative period was uneventful, and she was discharged home after 1 week.

Discussion

In 1900 the French physicians Klippel and Trénaunay described two patients who presented with a triad of varicose veins, port-wine stain and bony and soft tissue hypertrophy of an extremity. In 1918 Parkes-Weber described a patient with a similar triad and the additional finding of arteriovenous fistulae. Today, some authors use the term Klippel-Trénaunay-Weber syndrome to describe those patients who have clinically significant arteriovenous malformations as a component of their Klippel-Trénaunay syndrome. Other authors prefer to separate the original triad and the triad with the addition of arteriovenous malformations and use the term Parkes-Weber syndrome to describe this latter condition (1). Making the distinction is probably wise given the increased morbidity associated with arteriovenous malformations.

Today Klippel-Trénaunay syndrome (KTS) is defined as a combination of capillary cutaneous malformations, varicose veins or venous malformations and bony or soft tissue hypertrophy (1). Usually this triad is isolated to one extremity; however, multiple extremity, unilateral and even whole body involvement have been reported. While the leg is the most commonly affected site, the arms, trunk and rarely the head may also be involved. The syndrome affects males and females equally, has no racial predilection, and manifests at birth or during early infancy or childhood. The lower limb is the site

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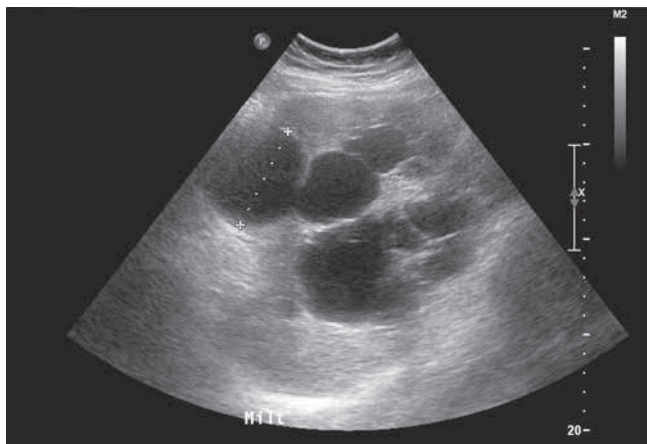


Fig. 1. — Abdominal US showed a massively enlarged spleen with several large anechoic cystic lesions.

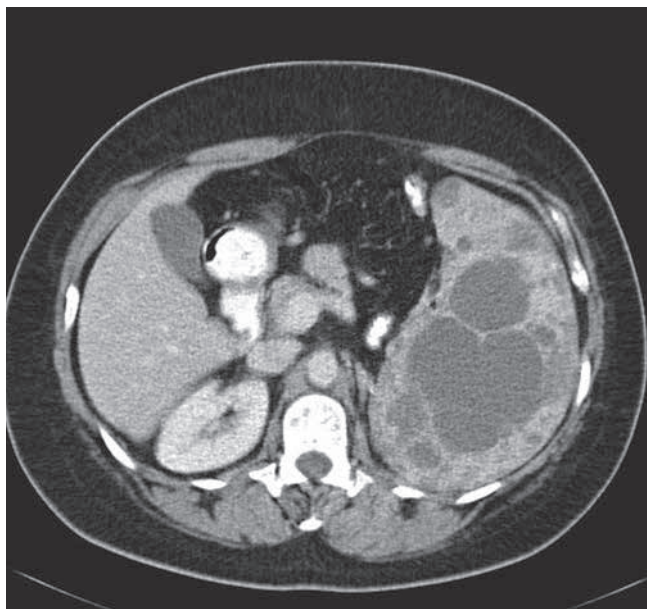


Fig. 2. — Venous phase contrast-enhanced abdominal CT showed an enlarged spleen with multiple large and several smaller cystic lesions. No contrast enhancing lesions were observed.

of malformations in approximately 95% of patients and lesions are usually limited to the skin (1).

Vascular malformations may involve other areas than the affected extremity and may vary in their depth of involvement as well as in their surface area distribution. Some vascular malformations extend to the muscle or bone of an affected extremity or even engage visceral organs. Visceral involvement in patients with KTS is rare, but has been described in the small bowel (2), colon (3), bladder (4), kidney (5), liver (6), spleen (7-10), mediastinum (11) and brain (12). When present visceral involvement can be a

significant cause of morbidity and mortality. In patients with gastrointestinal or genito-urinary tract involvement bleeding is the most reported symptom and this can range from occult blood loss to massive, life-threatening hemorrhages and consumptive coagulopathy. Splenic involvement in patients with KTS has been documented in the form of splenomegaly due to splenic vein stenosis (7), hemangiolymphangiomatosis (8), lymphangiomatosis (9) and hemangiomatosis (10). In our patient pathologic analysis showed that the observed splenic lesions were multiple cavernous hemangiomas.



Fig. 3. — No FDG-captation was observed in or surrounding any of the lesions on PET-CT examination. No lesions suggestive for primary neoplasm were observed. Notice the presence of the extensive vascular malformation in the left thigh.

In the general population splenic hemangiomas, although unusual, represent the most common benign primary tumor of the spleen with an overall incidence of 0.03%-14.5% based on autopsy findings (13). Splenic hemangiomas are usually less than 2 cm in size, only rarely attain a large size and may appear single or multiple. In KTS generally multiple lesions are observed. Hemangioma of the spleen usually has a silent clinical picture. However, large or multiple lesions causing significant splenomegaly may lead to fullness and left upper quadrant discomfort. At microscopic examination hemangiomas are characterized by an unencapsulated proliferation of dilated vascular channels that are lined with a single layer of endothelium and filled with red blood cells. The vascular channels that form splenic hemangiomas vary in size from capillary to cavernous. Microscopically, splenic hemangiomas are indistinguishable from hemangiomas

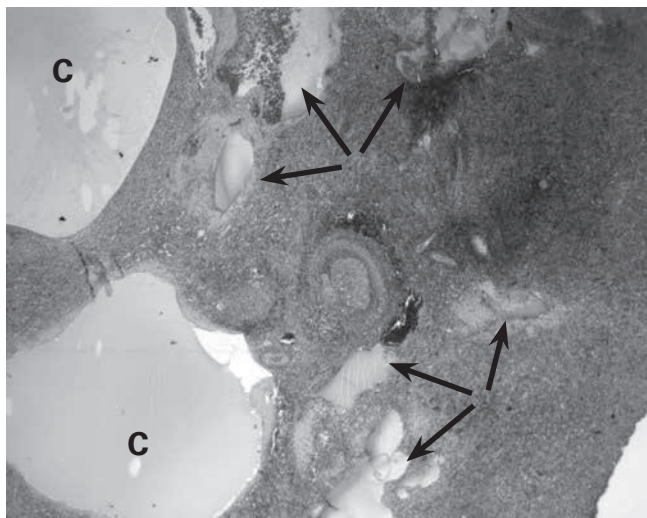


Fig. 4. — Histology of the spleen showed multiple large (C) and several smaller (arrows) cavernous hemangiomas lined by flattened endothelium and with a large number of erythrocytes in the lumen. No mitoses or dysplastic changes were noticed. A diagnosis of multiple cavernous hemangiomas of the spleen was made (hematoxylin and eosin staining with original magnification 4x objective).

in other areas of the body, such as the liver. However, cystic areas are more common in splenic hemangiomas due to cystic degeneration caused by central thrombosis and haemorrhage and may lead to atypical imaging characteristics (15). In these lesions CT may show some degree of peripheral enhancement; the cystic component does not enhance. In our patient no peripheral enhancement was observed.

Generally the prognosis for splenic hemangiomas is good and clinical follow-up does not usually include intervention. Spontaneous rupture with haemorrhage is a risk with larger (> 4 cm) lesions however, and rupture has been reported to occur in up to 25% of such cases (16). Rupture of splenic hemangiomas can occur either spontaneously or due to minor trauma. Recently Karakayali et al reported a case of spontaneous spleen rupture in a patient with KTS and diffuse splenic hemangiomatosis, illustrating that elective splenectomy may be indicated in these patients even when they are asymptomatic (17). Other complications of splenic hemangiomas include hypersplenism and Kasabach-Merritt syndrome. Malignant transformation has also been reported, but these cases may represent primary haemangiosarcoma.

Conclusion

KTS is a rare disorder with a wide variability of manifestations. Visceral manifestations are uncommon, but may probably not be as rare as previously believed as they may go unrecognized in asymptomatic or oligosymptomatic patients. In our patient diffuse hemangiomatosis of the spleen was only incidentally discovered during a routine physical examination. The atypical imaging characteristics of splenic hemangiomas are due to the more frequent cystic degeneration caused by central thrombosis and haemorrhage in splenic compared to hepatic hemangiomas. Elective splenectomy should be considered in patients with diffuse hemangiomatosis due to the increased risk of spontaneous or traumatic rupture.

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