

Gigantism of the lower limb in Klippel-Trenaunay syndrome: anatomy of the lateral marginal vein

Abdul-Rahman N R, Mohammad K F, Ibrahim S

ABSTRACT

The Klippel-Trenaunay syndrome is a combination of venous and capillary malformations associated with soft tissue and/or bony limb hypertrophy, with or without lymphatic malformations. Although persistent foetal veins are rare, the persistence of the lateral marginal vein is a common association in this syndrome. It results in venous hypertension, which gives rise to venous varicosities, which are commonly seen in this syndrome. This is a case report of a 28-year-old man with Klippel-Trenaunay syndrome, with persistence of the lateral marginal vein, affecting his right lower limb. He was treated with an above-knee amputation. The amputated limb was dissected to demonstrate the anatomy of the lateral marginal vein. To the best of the authors' knowledge, the gross anatomy of the lateral marginal vein has not been previously reported.

Keywords: Klippel-Trenaunay syndrome, lateral marginal vein, venous malformation

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INTRODUCTION

Klippel-Trenaunay syndrome is a rare condition. It consists of a combination of vascular malformations (capillary malformations, most commonly port-wine stains, venous malformations and varicose veins) and hypertrophy (bone and/or soft tissue).⁽¹⁾ There is a propensity to affect the lower limb unilaterally. It is usually not evident at birth and progresses slowly during growth.

CASE REPORT

A 28-year-old man presented with difficulty in walking due to gigantism of his right lower limb. He was diagnosed during childhood with Klippel-Trenaunay syndrome, and his right lower limb had progressively increased in size. His ability to walk had decreased, and standing for long periods of time had begun to cause him pain. Clinical examination revealed gigantism of



Fig. 1 Photograph shows gigantism of the right lower limb. There is a large port-wine stain, with a clearly defined border, along the lateral aspect. Superficial veins are dilated and there is a large varicosity in the lateral aspect of the right popliteal fossa.

his right lower limb. There was a large port-wine stain, with a clearly-defined border, along the lateral aspect. The superficial veins were dilated, and there was a large varicosity in the lateral aspect of the right popliteal fossa. The right foot showed changes consistent with lipodermatosclerosis (Fig. 1). Magnetic resonance (MR) imaging showed subcutaneous fat hypertrophy, diffusely infiltrated by small lymphovascular channels (Fig. 2). MR venography showed a large lateral marginal vein along the length of the right lower limb. There was an arteriovenous shunt at the knee draining into an ectatic venous sac (Fig. 3). He was treated with an above-knee amputation as the limb was cumbersome and interfered with his activities of daily living. The amputated limb was dissected to demonstrate the anatomy of the lateral marginal vein. The vein was dilated, valveless and

Department of
Orthopaedics
and Traumatology,
Faculty of Medicine,
Universiti
Kebangsaan
Malaysia,
Cheras,
Kuala Lumpur 56000,
Malaysia

Abdul-Rahman NR,
MBBCh
Orthopaedic Trainee

Mohammad KF,
MRCS, MSOrth
Orthopaedic Surgeon

Ibrahim S, FRCS,
MSOrth
Paediatric Orthopaedic
Surgeon

Correspondence to:
Dr Sharaf Ibrahim
Tel: (60) 3 9145 6007
Fax: (60) 3 9173 8609
Email: sharaf@ppukm.
ukm.my

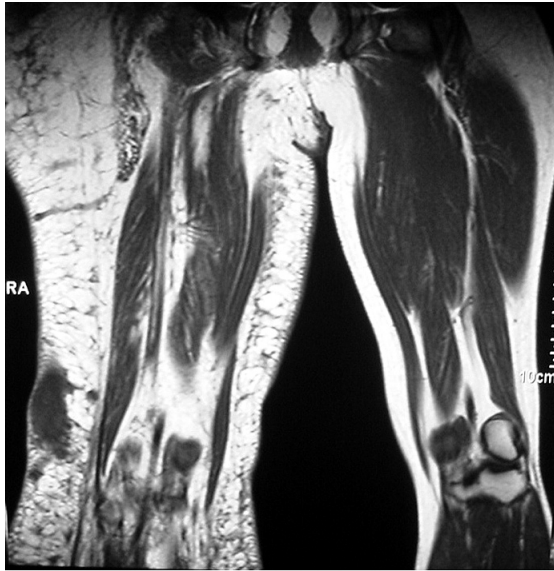


Fig. 2 Coronal T1-W MR image shows subcutaneous fat hypertrophy, diffusely infiltrated by small lymphovascular channels in the right thigh. The muscles are atrophied.



Fig. 3 MR venogram shows a large lateral superficial vein along the length of the right thigh (upper arrow). There is an arteriovenous shunt at the knee (lower arrow), draining into an ectatic venous sac.

thick-walled (Fig. 4). Histological examination of the subcutaneous tissues showed dilated lymphovascular channels within the thickened subcutaneous fat and atrophied skeletal muscles (Fig. 5).

DISCUSSION

The diagnosis of Klippel-Trenaunay syndrome is

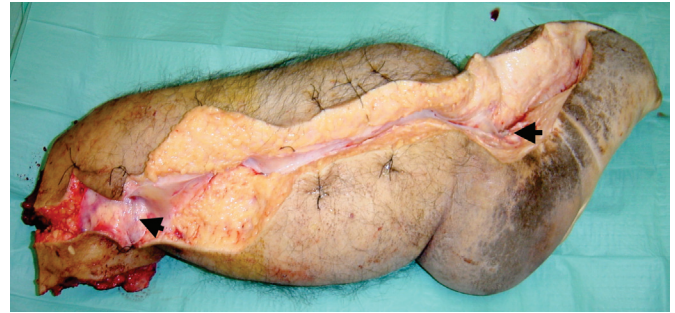


Fig. 4 Photograph of the dissected specimen of the amputated right lower limb shows the lateral marginal vein. This is a dilated, valveless, thick-walled superficial vein along its lateral length from the dorsolateral aspect of the foot (right arrow). Thickened subcutaneous fat surrounds the vein. A large sac in the popliteal fossa (left arrow) corresponds to the varicosity clinically and the venous sac on MR venography.

made based on the presence of at least two of the three cardinal features of capillary malformations, venous malformations and limb hypertrophy.^(1,3) Lymphatic malformations may be an association but do not necessarily constitute the syndrome.⁽¹⁾ They manifest as either lymphoedema or cutaneous lymphatic vesicles secondary to backflow from an obstructed or congested deep lymphatic system.

The port-wine stain is usually red to purple in colour and is flat. It usually has an irregular margin and a clear, sharp border but rarely crosses the midline.⁽¹⁾ It may or may not blanch on pressure. It usually occurs on the ipsilateral side as the affected limb. Thrombosis may cause depigmentation while other lesions may progress to additional changes, such as skin atrophy, bleeding or infection. Complete disappearance does not occur. It may be exacerbated by puberty and pregnancy. In large lesions, there is an increased blood volume, leading to shunting, and may cause high-output cardiac failure.⁽³⁾ Histologically, the capillary malformation is composed of vascular channels, lined by a single layer of endothelial cells, localised to the superficial dermis.⁽²⁾

Varicosities are extensive, atypically very large, take an erratic course and begin to manifest in early childhood.⁽¹⁾ Ulcerations may result in superficial thrombophlebitis, cellulitis or, in advanced, untreated cases, lipodermatosclerosis. Other venous anomalies include hypoplasia or aplasia, agenesis, duplication, valvular incompetence and aneurysmal dilatation of the deep venous system.⁽⁴⁾ The persistence of foetal veins is a rare congenital anomaly. They normally involute around the tenth to 12th week of intrauterine life.⁽⁵⁾ They are larger in diameter, thick-walled, frequently valveless and may not be visible. The commonest is the persistence of the lateral marginal vein, found in 68%–80% of Klippel-Trenaunay syndrome patients.^(1,6,7) It begins as a venous

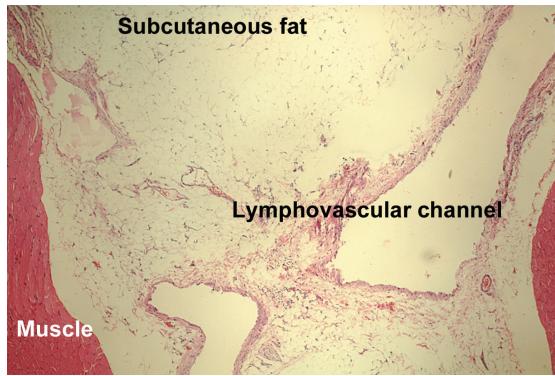


Fig. 5 Histological examination of the subcutaneous tissues from the amputated limb shows dilated lymphovascular channels within thickened subcutaneous fat and normal skeletal muscles (Haematoxylin & eosin, $\times 40$).

plexus on the dorsolateral aspect of the foot, taking a tortuous pathway proximally and laterally for a variable distance, before penetrating the deep venous system. Its termination may be single or multiple.⁽⁷⁾ In patients with deep venous aplasia, it acts as one of the collateral venous channels. It provides a source of venous reflux due to the absence of valves, and this can eventually cause persistent venous hypertension.

Chronic venous hypertension during childhood has been suggested as a cause of limb gigantism.⁽¹⁾ The persistent embryonic vein has also been postulated to augment blood flow, leading to an increased temperature of the limb, with resultant limb hypertrophy and gigantism. The increased limb girth is due to soft tissue hypertrophy, accompanied by increased vascular tissue thickness. The increased limb length is due to long-bone growth. The gigantism is disproportionately larger distally.

The best method of treatment depends on the extent of the lesion, the severity of the symptoms and the clinical condition of the patient. The first line of treatment of varicosities is supportive with elastic compression stockings. A percutaneous injection of sclerosing agents is an alternative method of treatment with small varicosities. The sclerosing agents irritate the endothelium, resulting in fibrosis and the obliteration of the vessel lumen. Surgical intervention is indicated when symptoms worsen. Venography is mandatory before surgery. If the deep venous system is dysplastic, an extensive varicectomy should not be performed because these varicose veins serve as substitute channels for the dysplastic deep veins.⁽¹⁾

In cases with normal deep veins, complete surgical resection of the marginal vein is the best form of treatment. This vein may have very large perforators to the deep

veins, and sometimes small arteriovenous fistulas may coexist. Closed stripping should be avoided because of the risk of excessive bleeding and haematomas. Adequate exposure of the veins is recommended, especially for large perforators to be ligated. In addition, large perforators in particular may be difficult to close and run the risk of the extension of thrombosis to the deep venous system. In cases of hypoplasia of the deep veins, the marginal vein can be resected because deep veins are able to dilate spontaneously to almost normal size after resection of the marginal vein. In cases of aplasia of the deep veins, the embryonic vein becomes a part of the main draining vessel of the limb, and resection is not possible. In some cases, arteriovenous fistulae to the vein increase the venous stasis of this valveless vessel significantly. In these selected cases, skeletonisation of the vein with ligation of all arterial inflow vessels can be performed.⁽⁸⁾

This case report illustrates the presence of all three cardinal features of Klippel-Trenaunay syndrome in the affected limb, with an associated persistent lateral marginal vein. In less severe cases, the syndrome is usually managed supportively and symptomatically. Amputation was performed in our patient as the large size of the limb had interfered with the activities of daily living. To the best of our knowledge, there is no previous report on the gross anatomy of the lateral marginal vein in Klippel-Trenaunay syndrome. Our report highlights the anatomy of the anomalous vein, which would be useful for surgeons considering surgical excision of the vein.

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