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# Clinical and Imaging Findings of Klippel-Trenaunay Syndrome: A Case Report

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**Abstract---***Klippel-Trenaunay syndrome (KTS) is characterized by the classic triad of capillary malformations manifesting as a "port-wine stain", venous varicosities, and bone and/or soft tissue hypertrophy. Imaging could help to diagnose and evaluation of KTS. Case Presentation: We report a 12-year-old with progressive enlargement of the right lower limb with geographic stain and venous varicosities. The patient also underwent MRI and angiography, which revealed multiple nidus with multiple feeding arteries. Conclusion: Klippel-Trenaunay Syndrome is a rare condition. MRI and Arteriography could help diagnose KTS for confirmation, monitoring progression, or identification of complications.*

**Keywords---***arteriography, imaging, intervention, klippel-trenaunay syndrome, MRI*

#### Introduction

Klippel-Trenaunay syndrome (KTS) is characterized by the classic triad of capillary malformations manifesting as a "port-wine stain", venous varicosities, and bone and/or soft tissue hypertrophy. This syndrome was described in 1900 by Maurice Klippel and Paul Trenaunay (Procaccini et al., 2021). Diagnosis of KTS is typically made when at least 2 of these 3 features exist. Patients with at least 2 of the 3 cardinal features have been classified as having an incomplete form of KTS. All three features are seen in up to 63 % of patients. The lower extremity is involved in approximately 95 % of patients. This disorder commonly affects unilateral extremity; however, it may involve different regions of the body and may involve other regions, such as musculoskeletal, craniospinal, head and neck, and visceral regions (Razek et al., 2019; El-Merhi et al., 2013). Imaging has an important role in the diagnosis and evaluation of KTS. Imaging is also mandatory to assess its severity and choice the treatment strategy (Procaccini et al., 2021; Wang et al., 2017). We present a case of KTS involving unilateral lower limb (Cohen & Weisskoff, 1991; Skorpil et al., 2004).

#### Case Report

A 12-year-old attended the surgical outpatient clinic at Dr. Soetomo General Academic Hospital with the chief complaints of progressive enlargement right lower limb for 2-year-old. He also complains about altered gait due to the discrepancy in limb length and getting worse. He had difficulty walking since 6-year-old. There is no history of trauma. From the physical examination, there is swelling, skin discoloration, and geographic stain due to venous varicosities in the right lower limb (Paramita et al., 2018; Macías & Martinez, 2019). There is also leg hypertrophy. There is no similar lesion on the opposite lower limb nor other body regions. The patient had an unenhanced and enhanced MRI of the right cruris with the result showed multiple nidus with irregular tortuous flow voids in proximal to mid cruris which appeared hypointense at T1WI/T2WI. The lesion extended to the muscular

compartment, subcutaneous, and skin causing soft tissue bulging around it. It also showed bone marrow intensity changes in tibia that hypointense at T1WI and hypo-to-iso intense at T2WI. The patient also had arteriography with the result of multiple wide arterio-venous dysplasia vascular lesions with multiple nidus at femoral regio to the right leg with feeding arteries from the branch of right superficial femoral artery, right popliteal artery, right peroneal artery, and right posterior tibialis artery. According to this arteriography appearance and supporting with MRI result and physical examination, the patient was diagnosed with Klippel- Trenaunay Syndrome (Fleming et al., 1991; Garg et al., 2000).



Figure 1. Clinical photograph of patient showing right lower limb hypertrophy, swelling, and venous varicosities (red arrow)

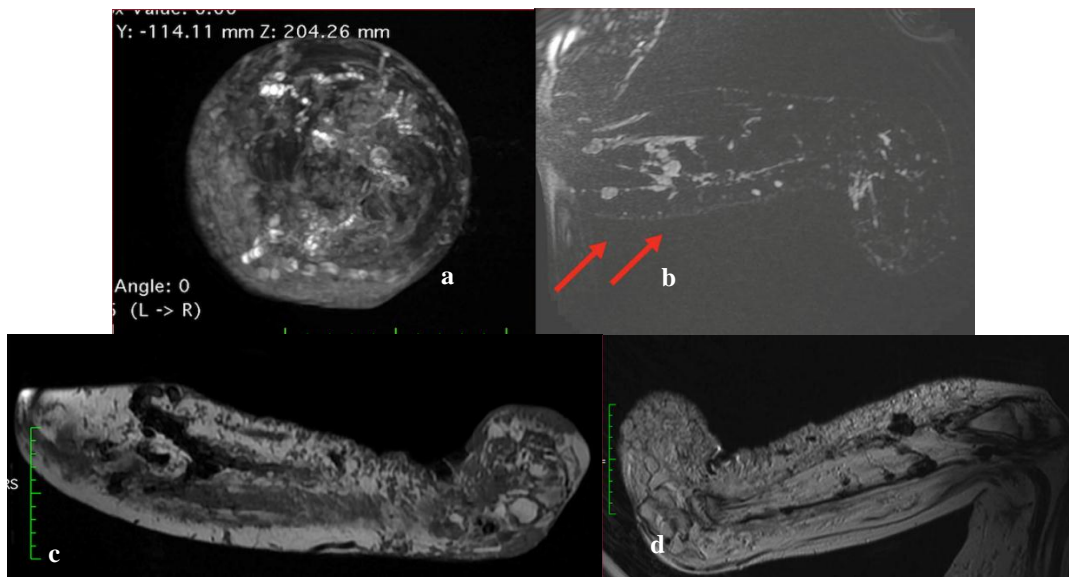
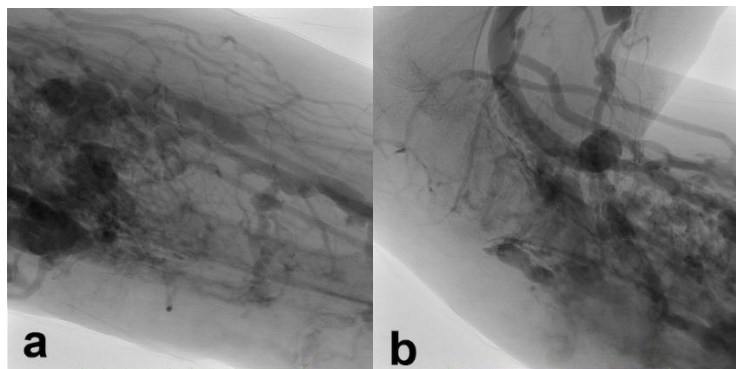


Figure 2. (a) MRI of right crus, Axial T1WI FatSat + C, (b) TRICKS, (c) Coronal T1WI, (d) Sagittal T2WI, showed multiple nidus (red arrow) with irregular tortuous flow voids in proximal to mid-right crus



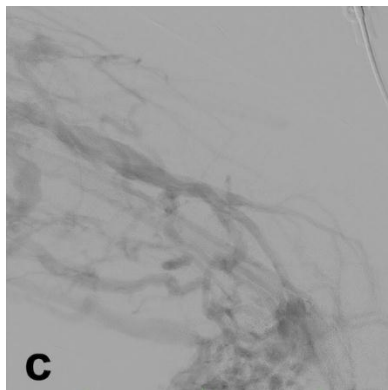


Figure 3. (a) Arteriography of right cruris, (b) Multiple nidus at femoral regio to the right leg with feeding arteries from the branch of right superficial femoral artery, right popliteal artery, right peroneal artery, (c) Right posterior tibialis artery

## Discussion

Klippel-Trenaunay Syndrome (KTS) also known by various names, including angio-osteohypertrophy syndrome or hemangiectatic hypertrophy is a rare congenital syndrome involving enlarged veins and arteries, limb hypertrophy, and capillary malformations. The incidence of KTS is approximately 2-5:100,000 live births with both males and females equally affected, although some studies proposed that KTS affects males more often than females, with no racial predilection, and usually manifests at birth, or during childhood (Reddy et al., 2015; Supekar et al., 2020). The etiology of KTS is unknown. Where as most patients demonstrate a normal karyotype, sporadic translocations have been reported of chromosomes 5 to 11 and 8 to 14; a supernumerary ringed chromosome 18 has also been described. Other theories have been postulated that include a list like paradominant inheritance pattern, somatic mosaicism of an otherwise dominant lethal gene embryonic disturbed vasculogenesis, and mesodermal defects, but none of them have been proved to have any definite association with the disease (Wang et al., 2017; Sharma et al., 2015).

Patients with KTS can have a wide spectrum of presentation from incomplete, mild forms of port-wine stains, and few varicose veins to severe disability, chronic venous insufficiency and lymphedema, massive limb overgrowths, limb length discrepancy, chronic pain syndrome, thromboembolism, recurrent rectal bleeding, and hematuria (Razek et al., 2019). Our case showed wine stain, varicose, and limb hypertrophy (McAuley et al., 2000; Møller et al., 2002). A port-wine stain is a cutaneous capillary malformation, which is present in 98 % of patients. The combination of low flow vascular abnormalities and lymphatic involvement makes the skin lesions appear bluish or purplish. The least common feature among all three findings is osseous and soft-tissue limb hypertrophy can affect both length and circumference of the extremity due to local hyperemia and venous stasis secondary to associated venous abnormalities. Varicose veins are almost a constant feature in patients with Klippel-Trenaunay syndrome. Bilateral involvement and truncal involvement are rare. However, extensive networks of venous malformation can extend to involve visceral organs of the pelvis and spinal structures (El-Merhi et al., 2013; Bertino et al., 2019).

The diagnosis of KTS is clinical and difficult to make even for the experienced physician as there is no true pathognomonic test. KTS should be initially evaluated with noninvasive imaging. The purpose of this is to determine the extent and severity of the disease which included the extent and distribution of structural and functional venous anomalies. Initial imaging workup for a patient presenting with the clinical triad of KTS is a Doppler ultrasound of the extremities for assessment of deep venous system involvement (Das et al., 2019; Alwalid et al., 2018). Plain radiographs are helpful for documentation of limb hypertrophy and to screen for limb-length discrepancies. Clinical measurement of limb-length discrepancies is generally limited and radiographic evaluation is necessary. A plain radiograph may also be used to search for phleboliths which are pathognomonic for venous malformations and can be a clue to prior hemorrhage or thrombus (Ochoco et al., 2019; Kharat et al., 2016). contrast-enhanced CT imaging with venous-phase protocols may be of particular use in pre interventional planning and cannot be avoided at times despite the best intentions (Wang et al., 2017).

In our case, the patient had undergone an MRI of cruris. MRI is both sensitive and specific for vascular anomalies and overgrowth and is regarded as the imaging of choice for evaluating most of the findings in KTS. MR imaging is the best technique for evaluating the extent of vascular malformations and their relationship with adjacent structures, which is critical for surgical planning (Wilson et al., 2001; Jacob et al., 1998). This can be seen on T2 SE sequences as multiple high signal foci within the muscles. Extension of the vascular malformations into the pelvis

may be identified. The role of MR angiography in analyzing vascular malformations in KTS has not been well defined, but the modality has the potential to depict these lesions with better accuracy. Precontrast and postcontrast MR imaging can be done to detect muscle hypertrophy, bone marrow changes of hypertrophied bone, and adjacent muscles and joints. Diffusion MR imaging can differentiate low-flow venolymphatic malformation from simulating lesions. Magnetic resonance venography can be used to delineate the abnormal venous drainage and as well as classification of an associated low-flow vascular formulation (Alwalid et al., 2018; Kharat et al., 2016).

After undergoing an MRI examination, our patient had underwent arteriography. DSA is the gold standard for confirmation of vascular malformations and arteriovenous fistulas. Arteriography is used to assess arterial malformations which may include abnormal origin, hypoplasia, dilatation, or stenosis (Clarke et al., 1995; Bakshi et al., 2008). Typical angiographic findings, which may also be seen on contrast-enhanced CT-scan or MRI, implicate lower limb superficial varicose drainage without opacification of the deep venous system. Conventional contrast venography is useful in assessing the route of venous drainage and also helpful in deciding treatment options like surgical resection or sclerotherapy (El-Merhi et al., 2013; Alwalid et al., 2018).

## Conclusion

Klippel-Trenaunay Syndrome is a rare case disease that should be diagnosed early with the classical triad and imaging modalities like MRI and Angiography are important as it helps in confirmation, monitoring progression, identification of complications, and intervention or surgical therapy.

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### Conflict of Interest

None

### Ethical Standard

None

### Department and Institution where work was done

The work was done in the Department of Radiology Dr. Soetomo General Academic Hospital, Surabaya, Indonesia

### Notes on patient consent case report

Written informed consent was obtained from the parent

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