

KLIPPEL–TRÉNAUNAY SYNDROME - A RARE ENTITY

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ABSTRACT

15 years old girl was presented to the plastic surgery department with soft tissue lesion of the right forearm since birth. There was an incidental finding of port wine stain on right chest wall. Her MRI shows low flow venous malformation of right antebrachial region with diffuse cutaneous, subcutaneous and intramuscular extension, varicose veins; Osseous and soft tissue hemi-hypertrophy with phleboliths was seen on forearm X-ray- the clinic-radiologic findings were in keeping with Klippel - Trénaunay syndrome. This report highlights the importance of radiology in the diagnosis of this rare entity in patients presenting with congenital vascular limb anomalies. **Keywords:** Klippel-Trenaunay syndrome, venous malformation, hemihypertrophy, MRI and MRV.

Introduction

Klippel-Trénaunay syndrome (KTS or KT), formerly Klippel-Trénaunay-Weber syndrome¹ and some-times angio osteohypertrophy syndrome and heman-giectatic hypertrophy,² is a rare congenital medical condition in which blood vessels and/or lymph vessels fail to form properly. It is characterized by a triad of capillary malformation (hemangioma or port-wine stain), venous varicosities, and bony or, soft-tissue hypertrophy.¹ The lower extremity is the site of involvement in approximately 95% of patients.² We present here this rare interesting syndrome in a 15 years old girl involving upper limb.

Case Summary

15 yr old female was referred to Civil hospital Karachi, with complains of swelling at right forearm since birth. Her parents had a consanguineous marriage. Rest of history was unremarkable. Her general physical examination showed upper limb length discrepancy and local examination revealed swelling over right anterior forearm which was soft and compressible,

nonpulsatile, mobile with bluish discoloration of skin. On right side of anterior chest there was bluish discoloration of skin noted. The clinical evaluation and Xray demonstrated limb length discrepancy. Her MRI of the forearm was planned. The imaging protocol included T1, T2 weighted, MRA, MRV, STIR and post contrast sequences. It demonstrated T1 hypointense and T2 lobulated hyperintense mass diffusely involving muscles, subcutaneous tissue and skin of hand and forearm with tortuous basilic and median vein.



Figure 1: Clinical examination reveals soft tissue lesion in right antebrachial region and an ill defined port wine stain on right side of chest (Reproduced with permission from the patient).

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Consequently this patient followup in plastic surgery department where her excision and coverage of lesion done and discharged uneventfully.



Figure 2: Radiograph of the forearm reveals limb length discrepancy with soft tissue and osseous hypertrophy (A) and multiple phleboliths in soft tissue lobulated lesion (arrows in B).

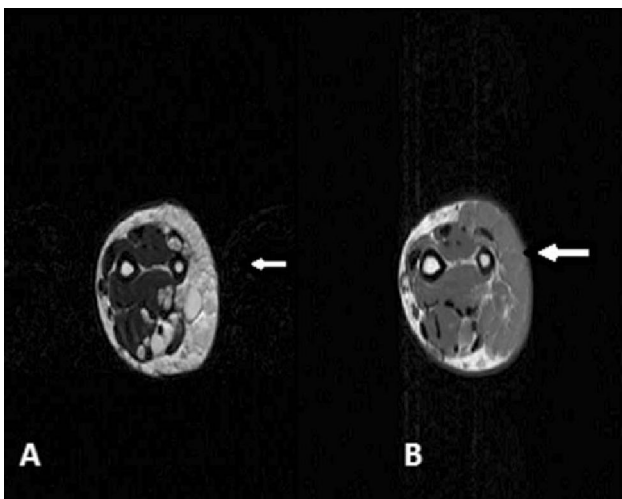


Figure 3: Contiguous axial T2W (A) and T1W (B) MRI images showing T2 hyperintense and T1 hypointense mass diffusely involving muscles, subcutaneous tissue and skin.

Discussion

Klippel-Trénaunay syndrome is a rare congenital syndrome of soft tissue venous malformation. Its complications include venous thromboembolism high-output heart failure gastrointestinal or genitourinary

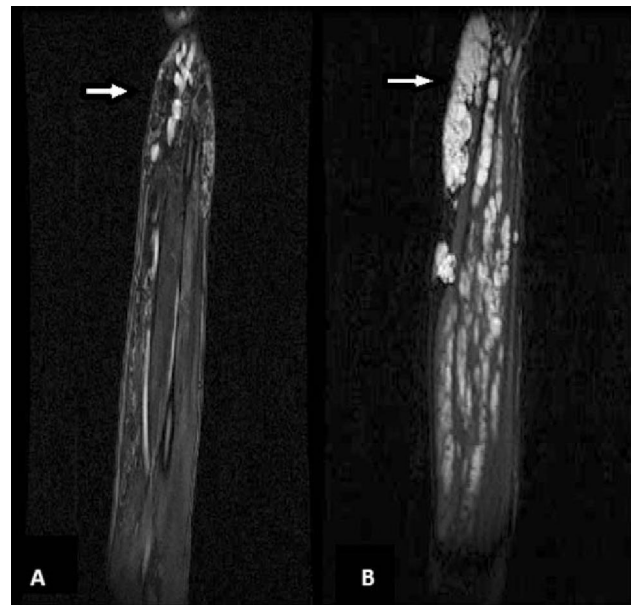


Figure 4: MRV (A) showing varicose veins in anterior intermuscular compartment of forearm and STIR image (B) showing lobulated hyperintense mass.

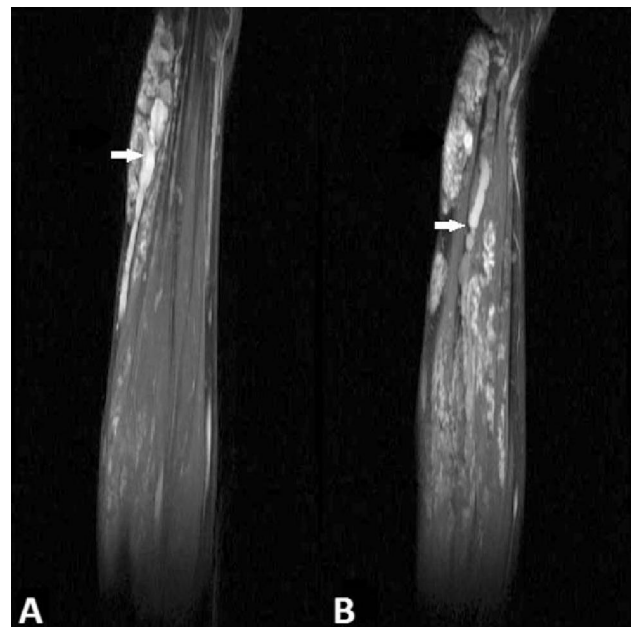


Figure 5: Post contrast images with heterogeneous enhancement of mass with tortuous basilic and median vein (arrows).

hemorrhage, if there is visceral involvement. If capillary malformations are large enough, they may sequester platelets, possibly leading to the Kasabach-Merritt syndrome (consumptive coagulopathy)³

A Prospective study of 20 patients was described by Kaddah RO et al 2011 who met the three criteria of KT syndrome. CT scanogram revealed leg length discrepancy (LLD) in all patients. MRI and MRV results

revealed different varieties of cavernous haemangioma and varicose veins, a form of surgical intervention was resorted to in 12 patients.⁴

Kharat AT et al in 2016 presented a 6-year-old child born out of nonconsanguineous marriage presented to us with a history of progressive enlargement of the right lower limb and port-wine stain on the lateral aspect of the thigh since birth. Magnetic resonance imaging (MRI) coronal images revealed diffuse irregular tortuous channels in the subcutaneous plane involving the entire thigh.⁵

Reddy OJ et al in 2008 45-year-old male patient whose right upper limb showed numerous soft bluish nodules with superficial ulcerations and varicosities extending onto right shoulder, right side of chest wall. There was soft tissue hypertrophy affecting the affected limb and there is difference in diameter of 3 cm between both arms at the level of 5 cm above olecranon process. The X-ray of affected limb shows multiple phleboliths.⁶

Akcali et al in 2008 described a case of a 14-year-old young woman who was a diagnosed case of KTS with associated multiple port-wine stain-type vascular anomalies and varicose veins involving the upper limb- compatible to our case.⁷

MRI with MRA/MRV are extremely helpful for the global evaluation of patients with KTS regarding confirmation of nature of the swelling as a vascular anomaly, as well as details about superficial and deep venous systems. Skeletal survey helps in detection of associated skeletal anomalies that necessitate correction.

The proposed radiology imaging protocol involved scanning by Skeletal survey, MRI, and MRA/MRV as well as baseline abdominal US for associated visceral involvements.

Conclusion

KTS represents a challenging problem in both diagnosis and management. Characterization of the different components of the syndrome is the area of challenge met by the radiologist. This study presents an example of a radiological protocol which may prove extremely beneficial in the diagnostic and surgical planning strategy.

References

1. James, William; Berger, Timothy; Elston, Dirk (2005). *Andrews' Diseases of the Skin: Clinical Dermatology* (10th ed.). Saunders. p. 585.
2. Kaddah RO, Lotfi U, Haggag M, Ghani HA. The role of radiology in the planning management of KlippelTrenaunay Syndrome (KTS). *The Egyptian Journal of Radiology and Nuclear Medicine*. 2011 Jun 30; **42(2)**: 201-10.
3. Mendiratta, V; Koranne, RV; Sardana, K; Hemal, U; Solanki, RS (2004). "Klippeltrenaunay Parkes-Weber syndrome". *Indian journal of dermatology, venereology and leprology*. **70(2)**: 119-22.
4. Kaddah RO, Lotfi U, Haggag M, Ghani HA. The role of radiology in the planning management of KlippelTrenaunay Syndrome (KTS). *The Egyptian Journal of Radiology and Nuclear Medicine*. 2011 Jun 30; **42(2)**: 201-10.
5. Kharat AT, Bhargava R, Bakshi V, Goyal A. Klippel-trenaunay syndrome: A case report with radiological review. *Medical Journal of Dr. DY Patil University*. Jul 2016; **9(4)**: 522.
6. Reddy OJ, Gafoor JA, Rajanikanth M, Prasad PO. Klippel-Trenaunay syndrome with review of literature. *Journal of Dr. NTR University of Health Sciences*. Apr 2015; **4(2)**: 120.
7. Akcali C, Inaloz S, Kirtak N. A case of Klippel - Trenaunay syndrome involving only upper limbs. *G ItalDermatolVenereol*. 2008; **143(4)**: 267-9.