Klippel-Trénaunay-Weber Syndrome: A Case Report.

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Abstracts: Klippel–Trénaunay–Weber syndrome is a syndrome combination of capillary malformations, soft-tissue or bone hypertrophy, and varicose veins or venous malformations. Most cases are considered sporadic. However, some of the possible differential diagnoses may have familial predispositions. We report one such case having right leg A-V malformation presented with complain of varicosity and non healing ulcer. [Arun G NJIRM 2016; 7(6):119-120]

Key Words: Klippel–Trénaunay–Weber syndrome, colour doppler, lower limb.

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Introduction: Klippel Trénaunay Weber syndrome (KTWS) and sometimes angioosteohypertrophy syndrome and hemangiectatic hypertrophy[1] is a rare congenital medical condition in which blood vessels and/or lymph vessels fail to form properly. The prevalence of KTWS (as such including Weber) is ~ 1 : 100,000.[2]

KTS classically comprises a triad of:

1) Port wine nevi
2) Bony or soft tissue hypertrophy of an extremity
3) Varicose veins or venous malformations of unusual distribution

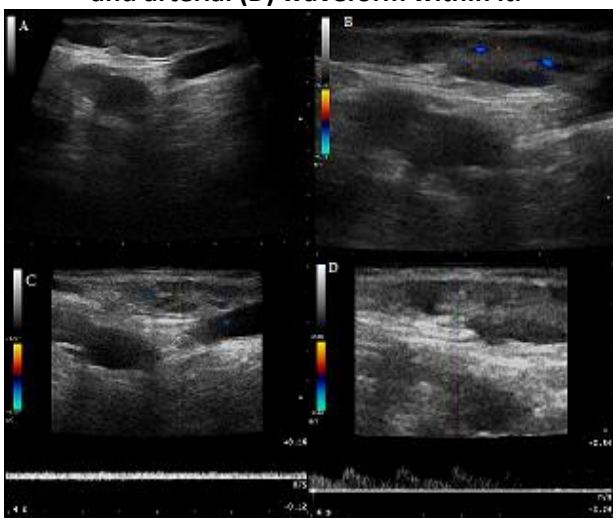
The diagnosis of KTS is usually made when any two of the three features are present[3].

Case report: A 25 years old female patient presented to Sir Takhatsinhji general hospital with the ulcer on lateral aspect of right ankle of 2-year duration.

Figure 1:(A) Port wine nevus over face, (B) Right lower limb hypertrophy, varicosity and port wine nevus

On examination there were port wine nevi over right lower and upper limb, right side of abdomen, and right side of face since birth(figure – 1A).The right lower limb was hypertrophied compare to left lower limb(figure – 1B).Varicosity noted at medial aspect of right lower limb(figure – 1B).

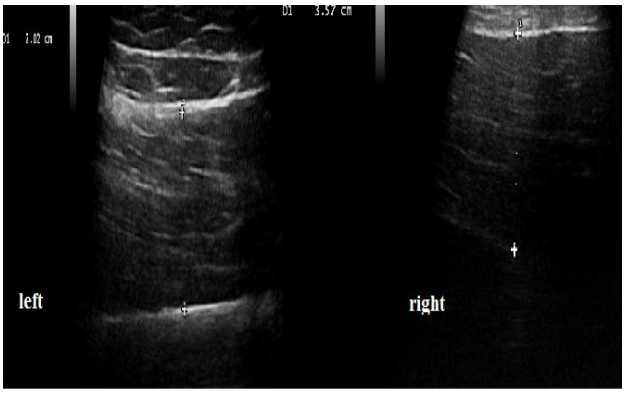
Figure 2: Rt. Thigh vascular malformation on grey scale (A) and color doppler (B) showing venous (C) and arterial (D) waveform within it.



Grey scale and color doppler sonography imaging of right lower limb:- Multiple heterogenous lesions with tubular anechoic structures within it noted at anterior and medial aspect of thigh(figure – 2A). The lesions show color flow on color doppler (figure – 2B) with both arterial and venous waveforms on spectral tracing (figure – 2C, D).Above description shows that the lesions are vascular malformations.

Right thigh muscles show hypertrophy compare to left thigh (figure - 3).Dilated and tortuous superficial veins noted over medial aspect of limb.

Figure 3: Right Thigh muscular hypertrophy compare to left.



Discussion: Klippel Trenaunay syndrome is characterized by a triad of port-wine stain, varicose veins, and bony - soft tissue hypertrophy involving an extremity.

Servelle believes that deep vein abnormalities, with resultant obstruction of venous flow, lead to venous hypertension, the development of varices, and limb hypertrophy[4].

Patients usually present in infancy. Features are often unilateral and typically affect one limb[5]. It may be diagnosed in utero[2].

Varicose veins result from damaged or defective valves in a vein. Vein gets damaged when the smooth muscle in the wall of vein weakens and the valves cannot support the weight of blood. Bone and soft tissue hypertrophy is a result of increased growth. In many cases, limb length is affected. In most cases, the girth of the limb is larger, although atrophy is seen in some patients. The lower limb is involved in about 95% of patients while upper limb involvement is seen in 5%[6].

On conventional radiography, bone elongation contributing to leg length discrepancy, soft-tissue thickening, or calcified phleboliths may be seen.

Prenatal ultrasound may diagnose KTWS as early as 15th week of gestation, based on limb hypertrophy and associated subcutaneous cystic lesions. 3D US may reveal leg width difference[2,7.]

Grey scale Usg and Colour doppler of limb shows multiple vascular malformations, varicosity, muscular hypertrophy of affected limb.

T2-weighted MR images may show malformed venous and lymphatic lesions as areas of high signal intensity. Typical angiographic findings, which may also be seen on contrast-enhanced CT-scan or MRI, include lower leg superficial varicoid drainage.

Conclusion: Occurrence of non-healing ulcer in addition to extensive port-wine staining of lower extremity, abdomen and face is a unique feature seen in our case.

The aim of this short report is to highlight the k- w syndrome, to avoid unnecessary and invasive investigations, and to avoid surgical complications.

A good history, clinical examination, basic sonography and colour doppler are sufficient enough to confirm a diagnosis of this entity in most cases.

References:

1. James, William; Berger, Timothy; Elston, Dirk (2005). Andrews' Diseases of the Skin: Clinical Dermatology (10th ed.). Saunders. p. 585.
2. Copel J. Obstetric Imaging, Elsevier Health Sciences. (2012)
3. Dhir L, Quinn AG. Persistent fetal vasculature and spontaneous hyphema in a patient with Klippel-Trénaunay-Weber syndrome. J AAPOS. 2010;14(2):190-2.
4. Servelle M. Klippel and Trenaunay's syndrome. 768 operated cases. Ann Surg Mar. 1985;201(3):365–73.
5. Cha SH, Romeo MA, Neutze JA. Visceral manifestations of Klippel-Trénaunay syndrome. Radiographics.25(6):1694-7doi:10.1148/rg.256055042
6. Phadke SR: Klippel Trenaunay syndrome. Atlas of Genetics and Cytogenetics in Oncology and Haematology, 2009;13(2):153-155
7. Marler JJ, Fishman SJ, Upton J et-al. Prenatal diagnosis of vascular anomalies. J. Pediatr. Surg. 2002;37 (3): 318-26.

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