International Journal of Biomedical And Advance Research

ISSN: 2229-3809 (Online)
Journal DOI:10.7439/ijbar
CODEN:IJBABN

Case Report

Klippel Trenaunay Weber Syndrome: A case report

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Abstract

Klippel Trenaunay Weber Syndrome (KTWS) is a rare, sporadic, complex manifestation characterised by micro arteriovenous and capillary malformation, along with varicose veins and bony and/or soft tissue hypertrophy, presenting usually in the infancy. Here a case of KTWS presenting in adolescence is reported.

Keywords: Parkes Weber Syndrome, port-wine stain, arterio-venous malformations, varicose veins, limb hypertrophy, Klippel Trenaunay Weber Syndrome.

1. Introduction

Klippel Trenaunay Weber Syndrome (KTWS) involves diffuse arteriovenous malformation (AVM) of a limb with overgrowth of that limb. The vascular malformations are usually limited to single extremity, though multiple extremities may be involved. The affected limb usually has cutaneous flush involving some of the skin, and is increased in length and girth.

2. Case Report

A 21 year old male presented with profusely bleeding ulcer over the dorsum of right foot. On examination, he had portwine stains and lipodermosclerosis over dorsum of his right foot and lower 2/3rd of the leg. Multiple discrete & grouped deep red to bluish black papules were present over the portwine stains. Localized gigantism of right leg was present along with dilated & tortuous superficial veins up to the level of mid-thigh (Figure 1).

On clinical examination, there was local rise of temperature of the right leg. Saphenofemoral valve was competent. Perforator incompetence was detected at mid-calf and above ankle region.

Patient told that his legs were normal till the age of 13 years, after which the right leg started increasing in thickness with appearance of those dark coloured patches and worm like swellings.

Arterial-venous Doppler showed varicosity of right great saphenous system & A-V communications in midleg & above ankle region. MR angiography revealed multiple A-V fistulae within right leg & proximal foot, draining into superficial & deep venous systems; and calibre of right common & external iliac arteries and all other arteries of right lower limb were larger than the left (Fig.2). 2D-echo showed mild pulmonary arterial hypertension with normal cardiac function. Patient was referred to Bangalore to vascular surgeon, where he underwent coil embolization of popliteal artery.

Figure 1: Anterior, medial and lateral view of the affected leg

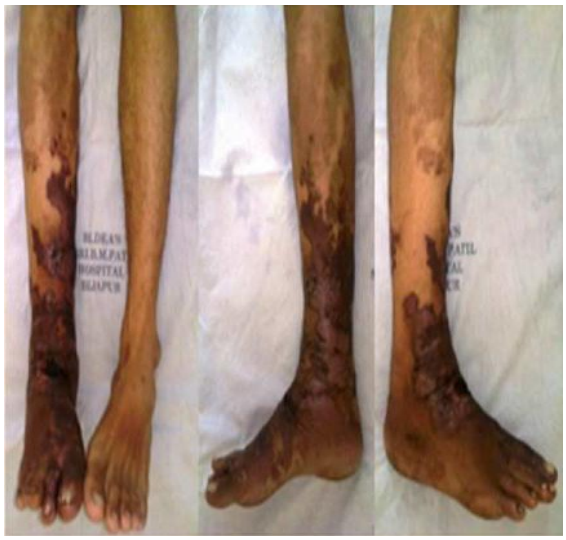
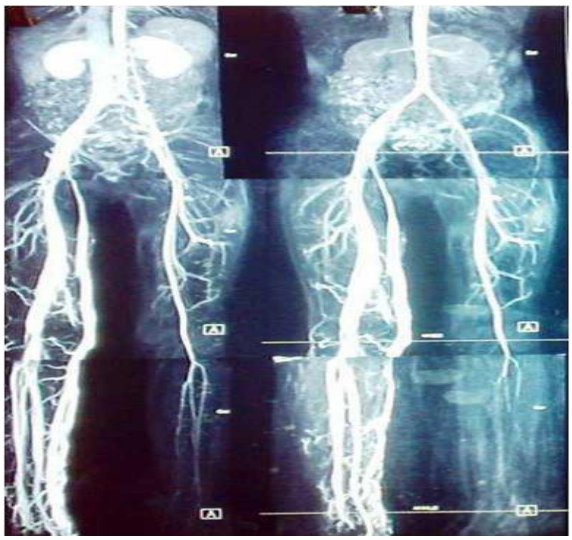


Figure 2: MR angiography showing increases calibre on right side



3. Discussion

KTWS is also known as Angio-Osteohypertrophy Syndrome, Congenital Dysplastic Angiectasia, Elephantiasis Congenita Angiomatosa and Osteohypertrophic Nevus Flammeus.[1] Legs are more often affected than arms.[2,3] When the amount of blood passing through the AVM is very large, an high-output cardiac failure can be observed.

3.1 Features

3.1.1. Vascular malformations

- Enlarged arteries and veins,
- Micro-arteriovenous malformations (AVM) (arteriovenous fistula),
- Atypical capillary malformations.

3.1.2. Soft tissue and skeletal hypertrophy of the affected limb.
3.1.3. Limb discoloration.
3.1.4. Warm skin on affected limb.

3.2 Aetiology

The aetiology of Klippel-Trenaunay Syndrome (KTS) is unknown. It is generally a sporadic disorder, although a paradominant inheritance pattern has been suggested.[4]It affects males more often than females. Incidence is about 2-5 per 1, 00,000. Some Parkes Weber syndromes are due to mutations in the gene RASA1 that encodes p120-Ras GTPaseactivating protein.[5]

3.3 Management

It can be helpful to wear compression stockings to prevent venous pooling in the affected extremity. Skin ulcers, infections and other skin problems can occur but usually the treatment is conservative. Management of large A-V fistulas can be done by embolization either by coil or gel foam.

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