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TRIAD OF TROUBLE: KLIPPEL–TRENAUNAY–WEBER SYNDROME – A CASE STUDY

General Medicine

Dr. G. Adarsh Junior Resident, Department of General Medicine, Great Eastern Medical School & Hospital, Srikakulam.

Dr. Usharani Budumuru Assistant Professor, Department of General Medicine, Great Eastern Medical School & Hospital, Srikakulam.

ABSTRACT

Klippel–Trenaunay–Weber Syndrome is a rare congenital vascular disorder characterized by capillary, lymphatic and venous malformations associated with soft-tissue and bone hypertrophy. The prevalence of the disease is approximately 1:100000 births. We report the case of a 29-year-old male presenting with chronic low back pain, limb hypertrophy and multiple venous abnormalities. Clinical evaluation revealed dilated tortuous veins, reddish-brown capillary haemangiomas, limb length discrepancy and facial port-wine stain. Imaging confirmed soft-tissue hypertrophy, dextroscoliosis, venous insufficiency with incompetent perforators. The patient was conservatively treated with analgesics, compression stockings and sclerotherapy was advised for venous anomalies.

KEYWORDS

Klippel-Trenaunay-Weber syndrome, vascular malformation, venous insufficiency, limb hypertrophy.

INTRODUCTION

Klippel–Trenaunay–Weber Syndrome is a congenital vascular malformation caused by somatic mutations in the PIK3CA gene, responsible for angiogenesis.Many other hypotheses have been postulated including translocations at (8;14) (q22.3;q13), supernumerary ring chromosome 18,terminal deletion 2q37.3, are some examples.It manifests as a triad of capillary malformations, venous insufficiency, lymphatic malformation and hypertrophy of soft tissue and bones of affected limb. It is a sporadic disorder without any racial or sex predilection.The prevalence is approximately 1 in 100,000.

CASE STUDY

A 29 year-old male presented with a history of low backache and multiple swellings over the left lower leg since 8 months and a port wine stain over face since childhood. Examination revealed multiple dilated tortuous veins from below the knee to the ankle along with few reddish-brown capillary haemangiomas and hypertrophy of the left lower limb associated with equinus deformity was noted. A diffuse flat reddish-purple coloured port-wine stain patch was also noted over the left side of the face.

Observations

The patient initially presented with chronic low backache, upon clinical examination multiple varicosities and capillary haemangiomas present over hypertrophied left lower limb. Clinical history revealed port wine stain over face since childhood raising suspicion for klippel trenaunay syndrome. The differential diagnosis are other similar syndromes like CLOVES, Proteus, as well as Diffuse capillary malformation with overgrowth(DCMO).

Management

Laboratory investigations revealed hemoglobin levels of 10g/dL. Imaging studies like Ultrasonography and color doppler of the left leg showed incompetent deep venous system with incompetent perforators and a patent saphenofemoral junction while X-ray of the left leg reveals soft tissue hypertrophy and calcifications. MRI of LS spine shows dextroscoliosis of spine with disc bulge at L4-L5 and L5-S1 levels.Ultrasound abdomen showed gross splenomegaly with multiple lobulated anechoic cysts suggestive of hemangiomas D-dimer levels were elevated (>10,000). The patient was conservatively treated with steroids, analgesics and supportive medication & advised to use compression stockings and sclerotherapy was advised for venous anomalies by the surgery team and now under regular follow up.

DISCUSSION

Klippel- Trenaunay- weber syndrome also known as Angio-osteohypertrophy syndrome is a rare sporadic disorder caused by post zygotic somatic mutation of PIK3CA gene which is responsible for angiogenesis. The disease is a classical triad of capillary, lymphatic and venous malformation with hypertrophy of soft tissue and bones of extremities mostly lower limb of affected side. Capillary manifestations are flat, red-coloured port wine stains seen in 90% cases. Venous malformations seen in 70 to 100% cases presenting with varicosities in superficial & deep venous systems, also in colon leading to deep venous thrombosis and intestinal bleed. Lymphatic malformations are seen in 15 to 60% cases with lymphedema. Musculoskeletal findings include soft tissue and bone hypertrophy leading to limb-length discrepancy.Imaging is an essential and sensitive diagnostic tool like color doppler ultrasound to assess vascular anomalies and an MRI to assess the extent of underlying malformations along with D-dimer levels are necessary. A multidisciplinary team approach involving orthopaedic limb correction, endovenous laser ablation or sclerotherapy, along with pulsed dye laser for port-wine stain is required. The treatment is mainly symptomatic and should be approached conservatively. For vascular and lymphatic malformations, sirolimus is an emerging treatment option.Complications like deep vein thrombosis, hemorrhage, pulmonary embolism, gastrointestinal bleeding, stasis dermatitis, cellulitis and limb hypertrophy are seen.

CONCLUSION

Klippel–Trenaunay–Weber Syndrome is a rare congenital vascular disorder with capillaro-lymphatico-venous malformations and soft tissue hypertrophy. We hereby present this rare and interesting case report from the southern part of India which provides review of clinical presentation. Timely diagnosis and co-ordinated multidisciplinary management along with regular follow-ups are essential to mitigate complications and enhance good patient outcomes.

Figures:

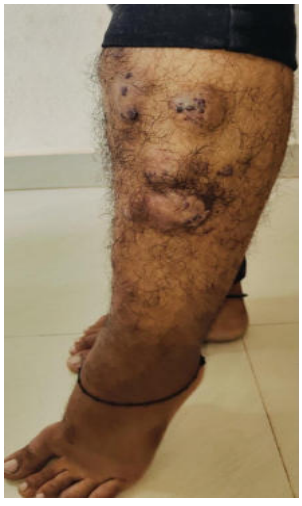

Figure 1: Capillary haemangiomas and varicose veins over hypertrophied left lower limb along with equinus deformity of foot.

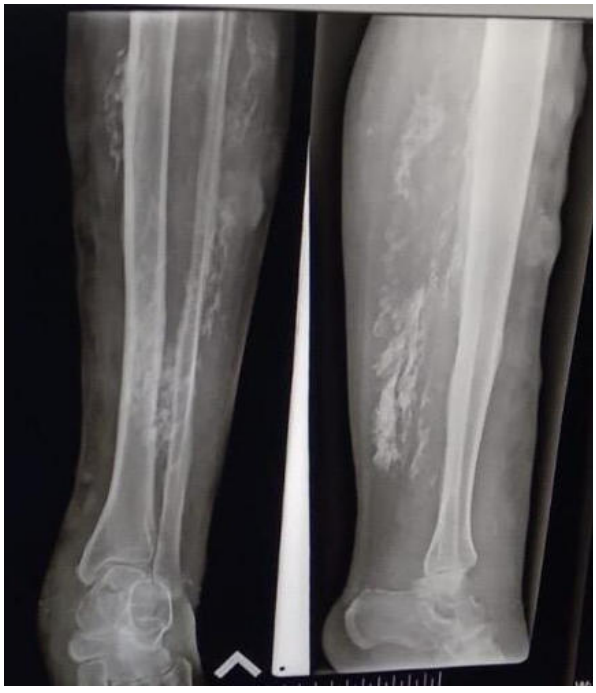

Figure 2: X-ray left leg showing soft tissue hypertrophy and calcification.

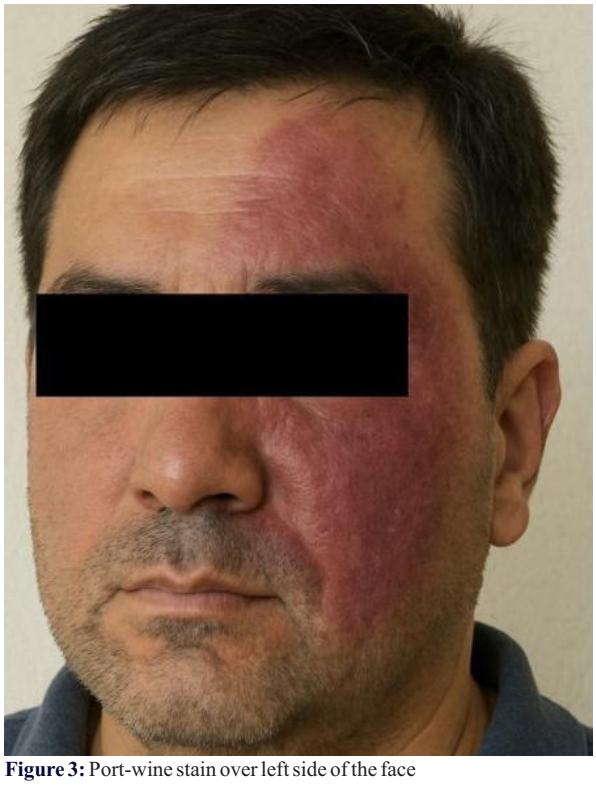

Figure 3: Port-wine stain over left side of the face

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