Klippel Trenaunay Weber (KTW) Syndrome with solid organ involvement: Case Report of a rare case

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Abstract—*Klippel-Trenaunay syndrome (KTW) is a rare, sporadic syndrome characterized by a triad of port-wine stain, varicose veins with or without venous malformations, and bony and soft tissue hypertrophy. It usually affects only one extremity, organ involvement is uncommon. A case of 17-year-old boy with this syndrome having solid viscous organ involvement was observed at surgery Outdoor. This case is rare so it was reported. Splenic lymphangiomatosis is also observed in this KTW syndrome case as association of AVM and lymphangioma together in spleen is rarest which was observed in this case. So it is a very rare case.*

Keywords: Arterio-Venous Malformations, Limb Hypertrophy, Port-Wine Stain, Splenic Lymphangiomatosis

I. INTRODUCTION

KTS syndrome was first described in 1900 by french physician Klippel and Trenaunay in two patients having port wine stain, varicosity in lower limb, bony and soft tissue hypertrophy of affected limbs[1]. In 1907, Parkes Weber described a patient with above mentioned features as well as an AVM of the affected limb. This was thus called Klippel Trenuanay Weber syndrome[2] It is a rare syndrome and rarely seen and reported by researches. When this KWT syndrome case observed in surgery outdoor, it was further evaluated and was found that it also have Splenic lymphangiomatosis which a further rare entity with KWT. So it was decided to report this rare case.

II. METHODOLOGY

A case report was framed and reported of a rare case of Klippel Trenuanay Weber syndrome case observed in surgery outdoor of hospital attached to Government Medical College, Kota (Raj) India. This case was admitted and was further evaluated on examination and investigation like CECT Abdomen, MRI Abdomen/Pelvis/Lower Limb shows dilated lymphatic channels in retroperitoneum, scrotum and all over left lower limb. Colour Doppler study of left lower limb was also done. Sigmoidoscopy and Gastroscopy of this case were also done for further evaluation. It was found that it also have Splenic lymphangiomatosis which a further rare entity with KWT. So this case falls in category of much rare case so it was decided to report this rare case.

III. CASE REPORT

A 17 year old male attended surgical OPD with huge lump abdomen and swelling in left lower limb. He was admitted and further interrogated in details. **On history** of the patient, salient history was revealed as follows:

1. A swelling at back was excised at the age of 4 day only, details of which were not available with the patients.
2. Surgical excision of lymphatic cyst in left lower limb was done in this case twice, one in 2002 and other one in 2007.
3. Patient had blood transfusions every 3 to 4 years for anaemia he had.

Now, he was admitted for huge lump abdomen and swelling in whole of left lower limb. Patient was examined in detailed as follows:-

Clinical examination:

Patient was well built except deformity of left leg and foot. Although he was neither cyanosis nor jaundice but had anaemia. He also did not had palpable lymph nodes. JVP was normal.

Abdominal examination revealed huge splenomegaly 16 cm below costal margin, firm, non tender and smooth with palpable splenic notch. No other organomegaly found. No free fluid in the abdomen. Scrotum was enlarged with worm like bag appearance on left side and extended upto lower abdominal wall. Testes and its cords were normal. (Fig-1)

Figure.1

Enlarged Scrotum with Worm like Bag Appearance

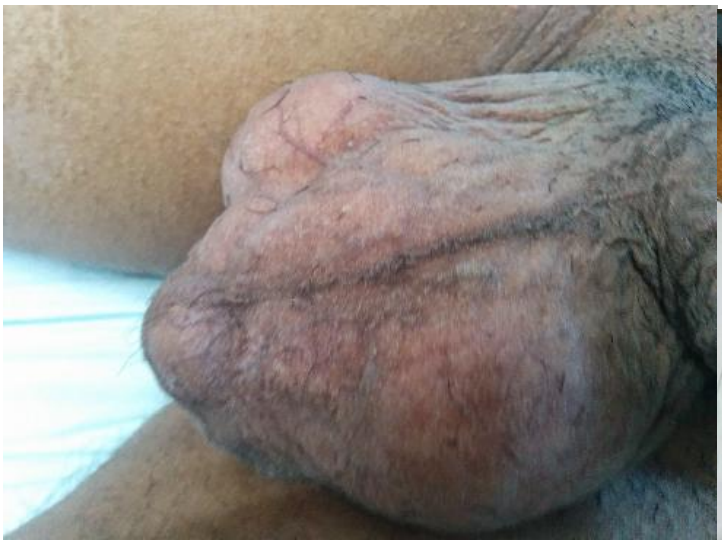
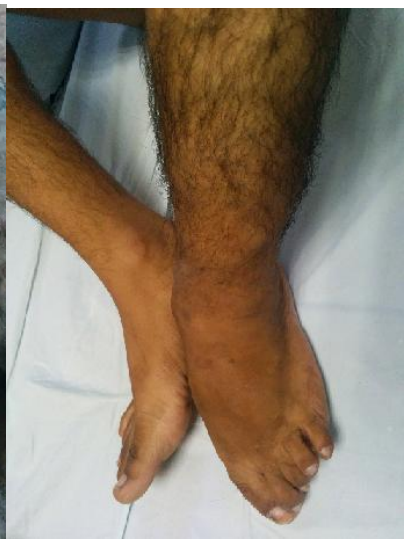


Figure.2

Medially deviated Foot with swelling



Left lower limb was deformed. There was huge swelling extending from thigh to feet with prominent veins and multiple scars over leg. Non pitting type oedema was there. Foot was deformed and medially deviated with swelling on dorsum of foot. Toes were greatly hypertrophied. (Fig-2)

PR examination and proctoscopy were normal and rest of systems examined and found normal.

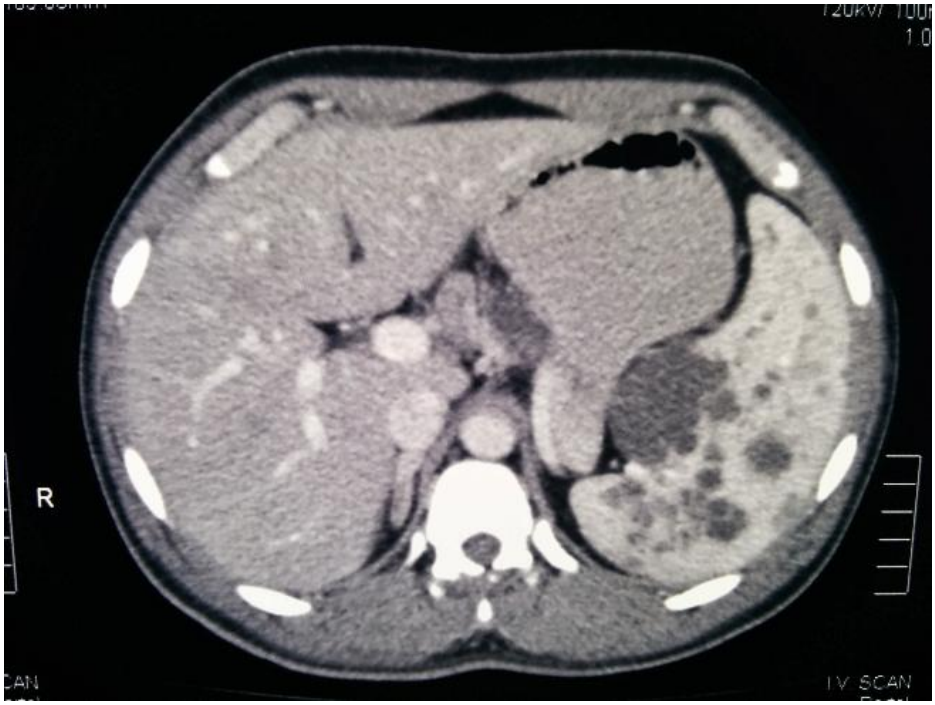
Investigations:- On investigations following were the salient findings:-

1. Hemoglobine (Hb) was found 6.4 gm,
2. PBF showed microcytic hypochromic anaemia,

3. LFT, RFT and serum electrolytes were normal.
4. Bone marrow examination normal,
5. PT/INR normal.
6. On radiological examination
    a. X-Ray foot (left) shows bony hypertrophy in toes. And X-Ray chest and abdomen were normal.
    b. USG Abdomen shows hugely enlarged spleen with multiple cystic areas over it and dilated fluid filled channels in retroperitoneum and portal cavernoma.
    c. On CECT:
        i. CECT Abdomen shows splenomegaly with multiple cysts all over suggestive of lymphangioma, dilated lymphatic channels in retroperitoneum, cysts also present in body of pancreas and liver. (Fig-3)
        ii. In CECT Skull no AVM seen intracranial.

Figure -3

CECT Abdomen showing Splenomegaly with Multiple Cysts



    d. MRI - Abdomen/Pelvis/Lower Limb shows dilated lymphatic channels in retroperitoneum, scrotum and all over left lower limb.

e. Color Doppler study of left lower limb showing varicose vein involving long saphenous, short saphenous systems and perforators, SFV incompetent.

7. Sigmoidoscopy reveals prominent veins in submucosa of rectum and sigmoid colon.
8. Gastroscopic findings were normal.
9. On laparotomy:
    a. Spleen was found hugely enlarged, adherent all around and multiple cysts of varying sizes present all over it. (Fig.4)
    b. Near the hilum of spleen, a large AVM was seen. (Fig.5)
    c. No spleneculi found. No other cyst could be seen over surface of liver/kidney/pancreas.
    d. Retroperitoneum was full of dilated lymphatic channels.
10. Histopathology of spleen showed extensive lymphangiomatosis with some areas of arteriovenous malformations and thrombosis

Figure.4

Hugely Enlarged Spleen with multiple Cysts

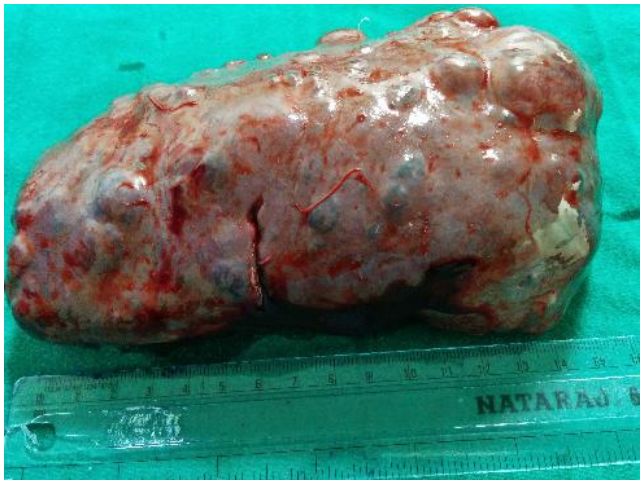
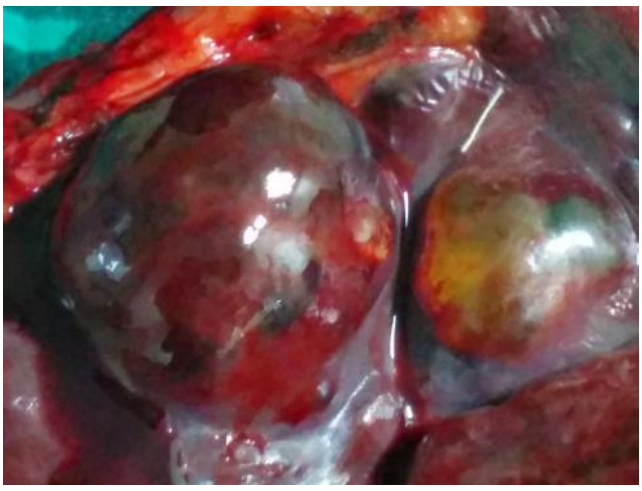


Figure.5

A large AVM near the hilum of Spleen



Management:-Patient was thus diagnosed as KTS and was planned for splenectomy owing to its huge size and its possible role in etiology of anaemia.

After blood transfusions, patient was taken for surgery and Laparotomy was done. Near the hilum of spleen, a large AVM was seen and carefully dissected and splenectomy was done. Dissected spleen was send to Pathology department for histo-pathological examination.

His post operative recovery was uneventful and patient was still under follow up.

IV. DISCUSSION

KTWS is estimated to affect at least one in 1, 00,000 people worldwide. It affects either sex equally and usually present at birth or infancy or childhood. The cause of KTS is yet unknown. Researchers suspect that the condition may result from changes in one or more genes that regulate the growth of blood

vessels during embryonic development. It is still unclear how blood vessel malformations are related to the overgrowth of bones and soft tissues.

A case report of KTWS in a monozygotic twin with an unaffected twin advances the theory of para-dominant inheritance pattern[3].Kihiczak et al reported that KTWS may result from a pathogenic gene for vascular and tissue overgrowth[4].

Malformations of veins are the third major feature of KTS. These abnormalities include varicose veins usually superficial but may occasionally involve deep veins and cause DVT. KTWS usually affect a single extremity although case of other limbs has been reported[5]. In the present case only left lower limb was grossly swollen and edematous.

Vascular malformations include capillary hemangioma, portwine stain, hemangioma in various part of body including gut and other viscous. Colonic AVM may cause hematochezia..

Other associated conditions include lymphatic obstruction, spina bifida, hypospadias, polydactyly and kasabach-merritt syndrome, a type of consumption coagulopathy.

Organ involvement is uncommon; solid viscous may be involved as in present case. Splenic lymphangiomatosis is seen but association of AVM and lymphangioma together in spleen is rarest as was noted in our case. Cause of anaemia in the present case can be chronic blood loss from gut or splenomegaly itself.

Diagnosis in such case is clinical and various imaging can confirm it. Investigations include haematological and bone marrow examination, USG abdomen, PCT skull, CECT abdomen, MR abdomen and limb and colour doppler study of lower limb. No definitive treatment is available and is to be managed for complication only.

CONFLICT OF INTEREST

None declared till now.

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