ORIGINAL RESEARCH PAPER

Volume-7 | Issue-5 | May-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

KLIPPEL–TRENAUNAY–WEBER SYNDROME PRESENTING AS RECURRENT RECTAL BLEED AND ANAEMIA: A RARE CASE REPORT

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ABSTRACT

Klippel Trenaunay Syndrome (KTS) is a rare congenital nonhereditary syndrome of vascular malformations and soft tissue and bone hypertrophy. Though the involvement of gastrointestinal tract (GI) is uncommon in KTS, it can be a source of life-threatening bleed with a high morbidity and mortality rates. We here report a case of a 26 year old gentleman, already a diagnosed case of KTS, who presented to us with recurrent GI bleed and anaemia necessitating blood transfusions every 20 days. This case report aims to highlight the management protocols for this rare presentation of the syndrome.

KEYWORDS

Klippel Trenaunay Syndrome, naevus vasculosus osteohypertrophicus, angioosteohypertrophy syndrome, hemangiectatic hypertrophy.

INTRODUCTION

Klippel-Trènaunay syndrome (KTS) characterized by a triad of :

1) Port-wine stain or "birthmark" (capillary malformations in the skin)
2) Soft tissue and bony hypertrophy (excessive growth of the soft tissue and/or bones) most often involving a single limb.
3) Vascular anomalies such as varicose veins.

Any 2 (out of the 3) in a patient are required for the patient to be labelled as a case of KTS.

The condition was first described by French physicians Maurice Klippel and Paul Trénaunay in 1900; they referred to it as naevus vasculosus osteohypertrophicus.(1)

The vascular malformations in KTS are reported to affect the GI tract, spleen, liver, heart, brain, kidney, bladder, penis, scrotum, vagina and vulva in addition to the limbs (limb hypertrophy and bony hypertrophy is usually seen in right lower limb).

CASE REPORT

A 26 years old male,a resident of Afghanistan, presented in our Surgical OPD with recurrent rectal bleed. He has been complaining of bleeding with stools since 1 year of age and had been taking treatment in the past from Afghanistan, Pakistan and India. Bleeding was usually associated with constipation. It was not associated with passing of stools. Initially, bleeding occurred every 2-3 days but gradually the frequency increased to daily bleeds. As the frequency and amount of bleeding increased over time, he started feeling giddiness and weakness and required blood transfusions every 3-4 months. Gradually, the frequency of blood transfusions increased to once a month and further to 3-4 units of packed red cell transfusions (PRC) every 20-25 days. For last 1 month he has been bleeding spontaneously per rectally. At the age of 16 years, he underwent Computed Tomography (CT) scan abdomen, angiography and colonoscopy at Pakistan and was found to have abnormal blood vessels in the colon and rectum. He underwent laser ablation for rectal varices and remained asymptomatic for 5 years.

In 2011, he underwent laser ablation for lower limb varicosities. The varicosities decreased in size but persisted.

At admission, his haemoglobin was 5.7 gm% ; rest all blood parameters were within normal limits, including the coagulation profile. On examination, he was pale but active. He had port wine stain on his right buttock **(FIG 1)** and right lower limb hypertrophy with varicosities (**FIG 2, 3).** He had no history of cardiac illness, hypertension, or diabetes.

His colonoscopy was planned in our hospital but because of poor preparation, was abandoned. CT angiography of abdomen showed marked circumferential thickening of rectosigmoid junction with wall calcification and multiple dilated tortuous venous channels in the mesorectal fascia till the anal verge. Mild caecal wall thickening **(FIG 4)**. In addition he had right sided varicocoele **(FIG 5)** and hypertrophy of gluteus soft tissue.

He was transfused 4 units of PRC preoperatively. Patient was counselled for abdomino perineal resection (APR) with permanent colostomy but he refused to give consent for permanent colostomy and hence was planned for Low Anterior Resection (LAR) with temporary ileostomy.

At surgery, the sigmoid colon and rectum showed multiple veins in the wall. Mesorectum was very vascular with thin walled varicosities (**FIG 6**). Sigmoid colon resection was done, without deep pelvic dissection, for it would have been impossible to avoid conversion to APR. Primary bowel anastomosis was performed along with a diverting ileostomy.

Grossly, the resected left colon and rectum showed extensive mucosal varicosities with nodular changes (**FIG 7**). The mucosa showed cobblestone appearance. The walls of resected specimen were thickened and bluish red in appearance **(FIG 8)**. The serosal surface showed markedly dilated veins and prominent varices.

Histology of resected specimen showed cavernous haemangioma involving the sigmoid colon and rectal wall transmurally with extension into mesorectum and sigmoid mesocolon and with focal mucosal ulceration. Lymph nodes in mesorectum showed reactive hyperplasia. Distal resected margin was positive of the lesion.

Postoperative course was complicated by intraabdominal bleed but was managed conservatively. He was transfused 15 units of PRCs & 10 units Fresh Frozen Plasma (FFP). He was discharged on 7th postoperative day in stable condition. At the time of discharge his haemoglobin was 9.7 gm% and was not bleeding per rectum.

He got readmitted 3 days after discharge with complaints of multiple episodes of vomiting which were nonbilious and non bloody in nature. Ileostomy was functioning well. He had decreased urine output. His haemoglobin was 12.1 gm/dl, blood urea 121mg/dl, serum creatinine-4 mg/dl and serum potassium 6.4 mEq/dl. He also complained of pain in right leg and thigh ; however venous doppler ultrasound of lower limb did not show evidence of DVT. He was managed conservatively and was discharged in stable condition with normal renal parameters.

Two months postoperative, he was doing well. He was not bleeding per rectum and his haemoglobin was 12 gm%.

He was planned for ileostomy closure after evaluation of the residual disease; however he was lost in follow up.

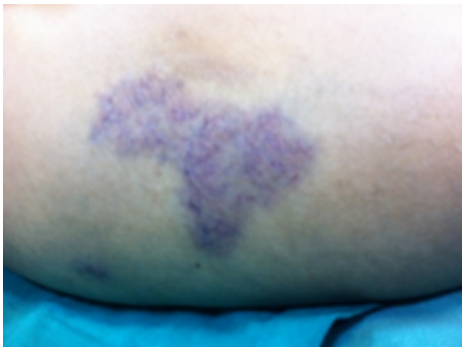

FIG 1 : Port wine stain in buttocks

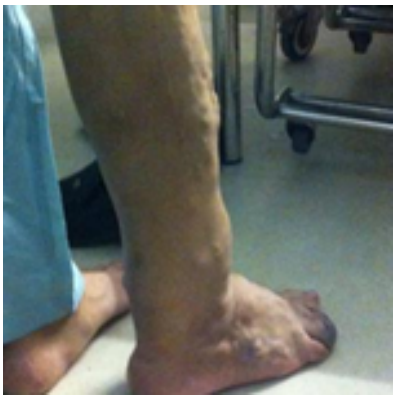

FIG 2 : Limb Hypertrophy and Varicosities

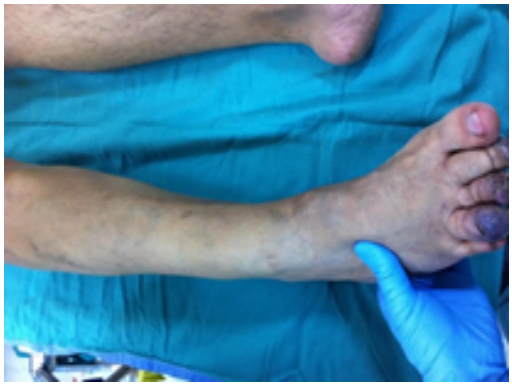

FIG 3 : Limb Hypertrophy and Varicosities

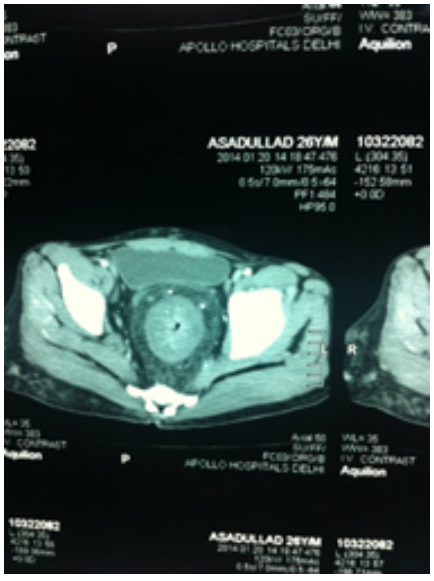

FIG 4 : CT scan showing thickened rectal wall with varicosities

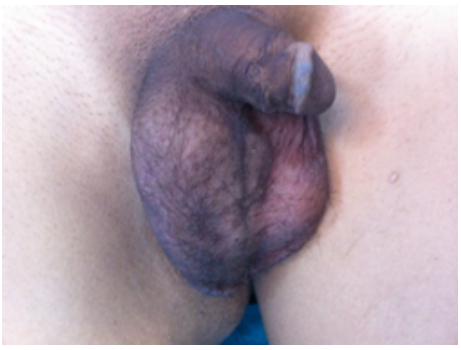

FIG 5 : Varicocoele Right side scrotum

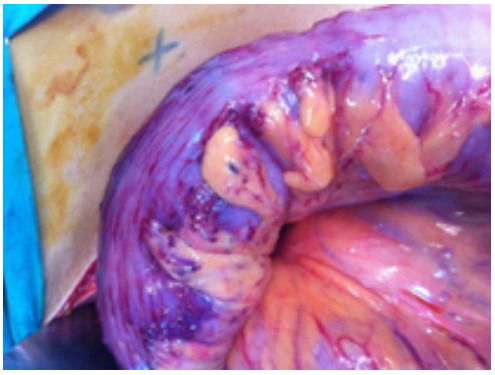

FIG 6 : Per operative intestinal wall thickenings

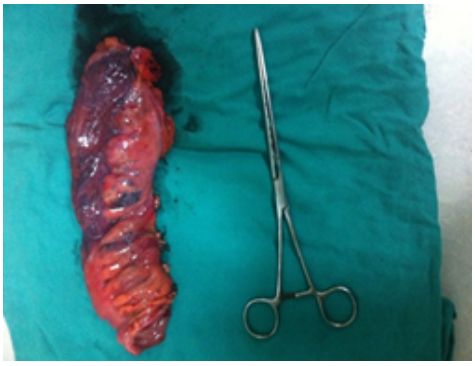

FIG 7 : Gross specimen

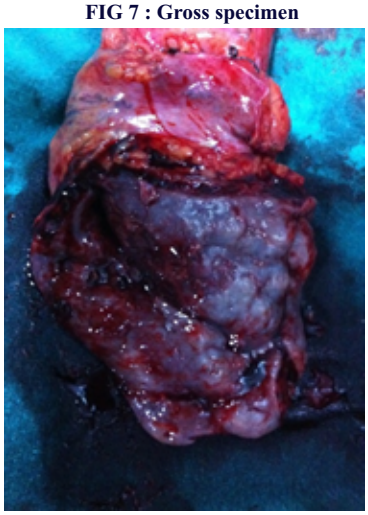

FIG 8 : Cut section of rectum showing venous malformation in the wall

DISCUSSION

Klippel–Trénaunay–Weber syndrome is also named as simply Klippel–Trénaunay syndrome(KTS) or sometimes angioosteohypertr ophy syndrome and hemangiectatic hypertrophy.

KTS is a nonhereditary congenital disorder affecting men and women equally. It is a rare condition, affecting about one in every 20,000 to 40,000 children. Children of all ethnic groups can be born with Klippel-Trenaunay syndrome.

Klippel-Trenaunay syndrome is present at birth but often the only visible sign in babies is the port wine stain. The diagnosis may not be confirmed until the varicose veins and limb hypertrophy become more noticeable.

What causes Klippel-Trenaunay syndrome is not known . There are various theories about possible causes, including problems with how the blood vessel system develops during pregnancy, but more research is needed to confirm the cause.

Few noteable personalities who suffered with this syndrome were
- Casey Martin , professional golfer
- Billy Corgan , lead singer for The Smashing Pumpkins
- Patience Hodgson , lead singer for The Grates
- Celeste Leon (Wisenhant) , JPS Family Medicine Resident

Most of the reported cases have dealt with the peripheral manifestations of KTS. However, it is now believed that intestinal involvement may be more common and may go unrecognized in patients without overt GI symptoms (**2**).

Visceral hemangiomas in KTS have been described involving organs such as the GI tract, liver, spleen, bladder, kidney, lung, and heart (**3, 4**). The commonest reported cause of GI bleeding is attributed to diffuse cavernous hemangiomas of the distal colon and rectum, found in an estimated 1–12.5% of KTS cases **(4, 5, 6, 7, 8, 9, 10, 11).** There have been reports of bleeding from jejunal hemangiomas but are rare **(2)**. Oesophageal varices secondary to prehepatic portal hypertension from cavernous transformation or hypoplasia of portal vein can be another significant source of GI bleeding **(11, 12)**. However, vascular malformations affecting the whole of GI tract are rare **(13).**

In one of the largest published series of KTS patients, hematochezia was reported in only six of 588 patients, although a few other cases may have gone unnoticed. Clinical manifestations range from occult to massive, life-threatening hemorrhage .Vascular malformations involving the GI tract have been a source of significant morbidity and even mortality (**5**, **14**). KTS patients with clinically significant hemorrhage usually require resection of the involved bowel segment (**14**).

Though bleeding is the most common manifestation of hemangiomas involving the GI tract, they may also present with ascites and bowel obstruction if there are giant hemangiomas involving the retroperitoneal space ; it is rare **(15)**.

The nature and extent of GI involvement are best diagnosed by endoscopy and selective visceral angiography. However, in atypical presentation of KTS, colour Doppler ultrasound and duplex scanning are very useful to establish the diagnosis **(16)**. CT scan and magnetic resonance imaging (MRI) can assess the intraabdomianal hemangiomas and their extension in abdomen and pelvis which helps in assessing the response of treatment and prognosis **(17,18).**

The management of intestinal involvement in KTS depends on extent and severity of bleeding. Although conservative management and iron supplements for iron deficiency anaemia may be sufficient in the patient who present with occasionally non significant bleeding, it is unlikely to be effective in the long term in view of the diffuse and progressive disease process .Transfusion dependent, life threatening bleeding episodes and poor quality of life require definitive therapy **(15).**

Endoscopic therapy either alone or in conjunction with surgery has been reported in the management of KTS patients with colonic hemangiomas. Photocoagulation using the argon laser was successful in treating a hemangioma involving the distal 7 cm of the anorectum in a boy with KTS **(6)**.

In another report, the neodymium:yttrium-aluminum-garnet laser was successful at treating residual lesions after partial colectomy for visceral hemangiomas in two patients **(7)**.

Endoscopic laser therapy, however, has a limited role in KTS patients and is best reserved for management of localized lesions or ablation of postoperative residual disease. The argon plasma coagulator may be a more efficient and less costly tool compared to the above-mentioned laser modalities, although its use in this condition has not yet been reported. Photodynamic therapy (PDT) using a low-dose photosensitizer has shown promise in ablating radiation-induced angioectasias within the rectosigmoid with minimal tissue necrosis **(19)**. Its ease of application and its capability of ablating vascular lesions in a circumferential fashion make PDT an appealing technique in treating diffuse hemangiomatosis. PDT has yet to be evaluated in this setting.

Rectal hemangiomatosis has usually been treated surgically by means of an abdominoperineal resection of the involved colon with a permanent colostomy **(4, 14, 20)**. In KTS, surgery is challenging because of the extensive visceral involvement making an attempt at sphincter-saving procedures even more daunting. However, because patients are often young adults, proctectomy with restoration of anal function by coloanal anastomosis is preferable **(30)**. Radiotherapy may prove suitable for these diffuse lesions, but data are lacking **(4, 22)**. Angiography usually serves its purpose as a diagnostic procedure, but angiographic embolization may be useful if a specific active bleeding site is encountered.

Telander et al. have reported in successfully treating a patient with segmental colonic resection, rectal mucosectomy and coloanal anastomosis in patients with recurrent rectal bleeding **(23).**

Reports are there where people have even resected the bowel laparoscopically and given good results **(24).**

CONCLUSION:

This is the first case of KTS we have encountered. We went through the literature extensively before we took up the case. The main problem which we faced during surgery was the highly vascular nature of the lesion. Any attempt to dissect the colon or rectum in the pelvis led to profuse bleeding which required immense patience to control. Even though we controlled the bleeding well before closing the abdomen, patient had massive haemoperitoneum necessitating 25 units of blood products. Our experiences with this very case defogged further the "difficult to treat" nature of this very syndrome. We feel, the surgical management options for distal gut lesions may be resection and primary anastomosis or resection coupled to permanent diversion but the most important factor to influence the decision of choosing amongst the two will be the pre requisite that **the margins of the resected specimen should be clear of the disease.**

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