KLIPPEL-TRENAUNAY SYNDROME: A RARE CAUSE OF INTERMITTENT LOWER GASTROINTESTINAL BLEEDING

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ABSTRACT

Klippel-Trenaunay syndrome is a rare non-hereditary congenital abnormality characterized by a clinical triad of soft tissue hypertrophy of extremity, varicosities and cutaneous hemangiomas or lymphongioma. Bleeding per rectum is an uncommon but potentially serious complication. We herein report a case admitted to the gastroenterology ward Postgraduate Medical Institute, Lady Reading Hospital Peshawar, with intermittent haematocezia and symptomatic iron deficiency anemia .The patient did not seek any help for gastrointestinal bleeding until his admission to our unit. He was also having gigantism of his right leg and port wine angiomatous spots on his buttocks and leg. The literature on the evaluation and management on this case is reviewed.

***Key Words**: Klippel-Trenaunay syndrome, gastrointestinal bleeding, soft tissue hypertrophy, venous varicosity.*

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INTRODUCTION

Kippel-Trenaunay-Weber syndrome is a rare congenital syndrome involving enlarged veins and arteries, limb hypertrophy and capillary malformations. In 1900, French physicians, Klippel and Trenaunay, first described what became known as Klippel-Trenaunay syndrome after two patients presented with a triad of symptoms — port-wine stain, varicose veins, and bony and soft tissue hypertrophy involving an extremity.

Later in 1907, Parkes Weber, who was unaware of Kippel and Trenaunay's earlier work, described a patient who also presented with these three symptoms as well as an arteriovenous malformation of an affected extremity, hence the condition became known as Klippel-Trenaunay-Weber syndrome[1-3].

Kippel-Trenaunay-Weber syndrome affects female and male equally at birth or during early infancy/childhood. The cause is unknown. Although there are some genetic studies underway, no specific genes that may cause Klippel-Trenaunay-Weber syndrome have been identified to date[4].

Vascular malformation involving gastrointestinal tract (GIT) have been reported and can be a source of significant morbidity and mortality. Visceral hemangiomas in Kippel-Trenaunay-Weber syndrome may involve organs such as the Gastrointestinal tract, liver, spleen, Urinary bladder, kidney, lung and heart[5]. We report here a case of a young male presenting with intermittent bleeding per rectum and iron deficiency anemia with past history of excision of vascular malformation from right calf muscle. Physical examination along with colonoscopic findings confirmed the diagnosis of Kippel-Trenaunay-Weber syndrome.

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CASE REPORT

A 17 year old boy presented to the gastroenterology Department with history of bleeding Per-rectum, lower limb swelling on the right side and palpable bluish-black skin lesion on the same limb. The patient father gave history of repeated episodes of fresh bleeding per rectum since the child was 3 months old along with multiple blood transfusions since childhood.

The patient also developed skin lesion at the same time on the right lower limb, initially pinkish to reddish in color and smaller in size that gradually enlarged and became bluish-black in

color. The right lower limb gradually enlarged in size since childhood and there is history of surgery on the right lower limb when he was 4 years old. However the record of surgery was not available.

On examination, he had severe pallor. His pulse rate was 110 per minute, and blood pressure was 104/70 mm Hg. He was afebrile, and did not have icterus, cyanosis, clubbing or lymphadenopathy. The skin of the right lower limb was thick and hyperpigmented. There were multiple discrete and grouped deep red to bluish black papules and nodules on the anterior and medial aspect of right leg, anterior and lateral aspect of right thigh and right buttock. A scar of previous surgery was noted on the medial aspect of right leg. The right lower limb was larger and longer than the left, the dimensions were as follows.

		RIGHT	LEFT
THIGH:	Length:	51 cm	48cm
	Girth:	37 cm (mid thigh)	35cm (mid thigh)
LEG:	Length:	38 cm	35cm
	Girth:	28cm (10cm below tibial tiberosity)	21cm

There was no hepatosplenomegaly, chest was clear, cardiac sounds were normal and no abnormality on neurological examination.

On investigation, his hemoglobin was 6.0 g/dL, with hypochromic, microcytic anemia and normal platelets count. Prothrombine time, activated thromboplastine time, Bleeding time and Clotting time were within normal limits. Liver function test and Renal function test were normal.

Figure 1: Klippel Trénaunay Syndrome with Soft Tissue Hypertrophy of right leg.

Figure 2: Bluish black (port wine stain) on Right thigh

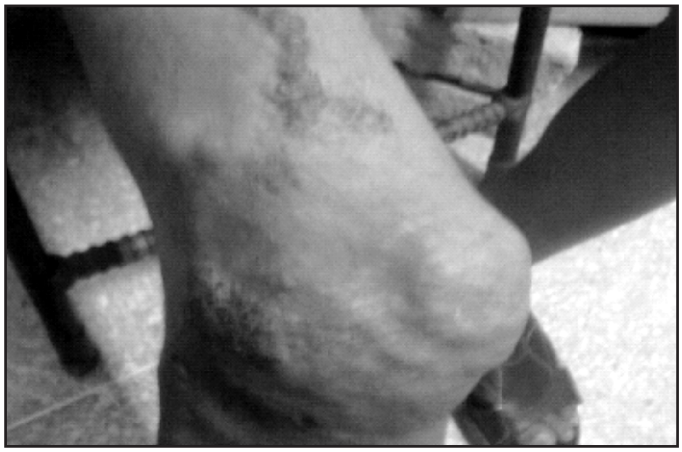


Figure 3: Localized Gigantism of right lower Lamb with Bowing

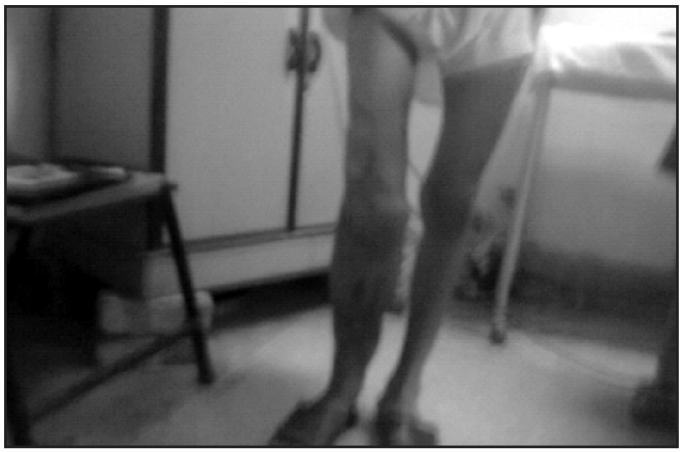


Figure 4: Surgical scar right leg

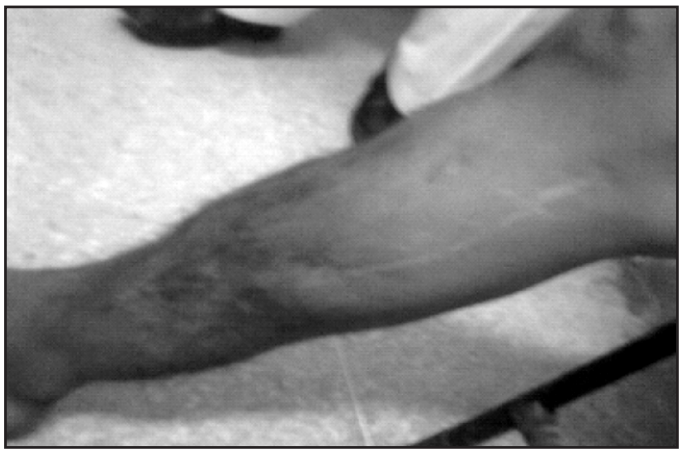


Figure 5: Vascular malformation Right arm

Figure 6: Perianal whitish-red nodules (vascular malformation)

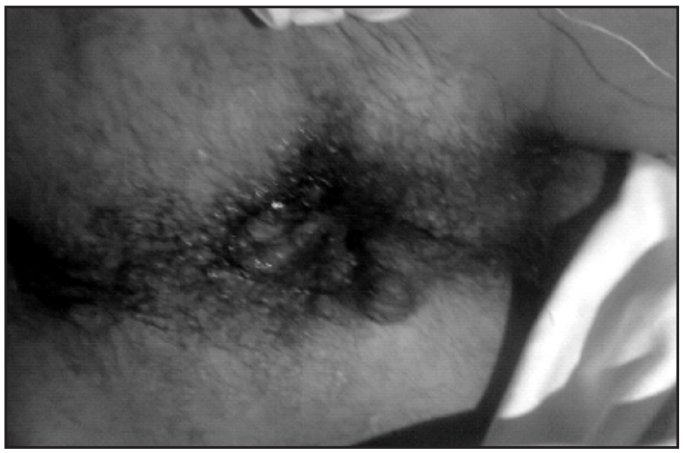
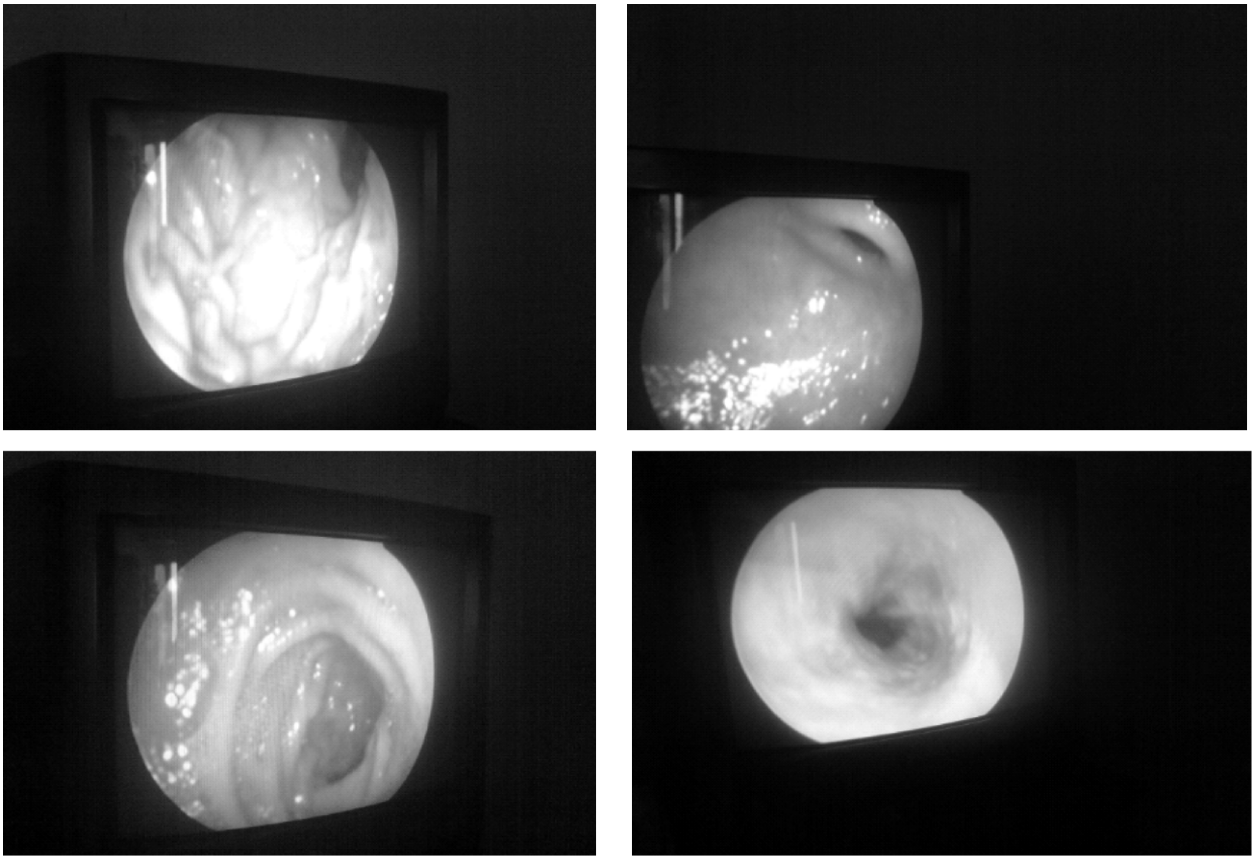


Figure 7: a,b,c,d: Colonoscopy picture showing Ulceration and Venous Varicosity.



Electrocariography and X-ray chest were normal. X-ray comparison of both lower limbs showed soft tissue hypertrophy of the right lower limb but no bony hypertrophy. Ultrasound abdomen and Doppler Ultrasound of lower limb was normal. Diagnostic colonoscopy revealed presence of numerous severely dilated and engorged blood vessels and mucosal ulcerations in the rectum and distal sigmoid colon. There were venous varicosities at the junction of normal and ulcerated

mucosa 40cm from the anal verge. Rest of the colonic mucosa was normal up to cecum. Pre-colonoscopic examination of the perianal area revealed vascular malformations in the form of whitish-reddish nodules.

Based on the vascular malformations in Gastrointestinal tract, cutaneous vascular malformation and the enlarged right leg, the patient was diagnosed as having Klippel Trénaunay Syndrome.

DISCUSSION

The case reported here, presented with characteristic features of Kippel-Trenaunay-Weber syndrome like soft tissue hypertrophy of the right lower limb and cutanouse angiocaratomas and bleeding per rectum secondary to varicose veins and ulceration in the distal 40cm of the large gut. Kippel-Trenaunay-Weber syndrome is a rare congenital disease with a reported incidence of 2-5/1,00,000 population. Kippel-Trenaunay-Weber syndrome patients presenting with Gastrointestinal bleed is also an uncommon presentation. Kippel-Trenaunay-Weber syndrome with Gastrointestinal involment is a progressive disease & may have life threatening bleeds occurring in perhaps as many as 20% of patients[6,7]. The local review showed that this is the first case of Kippel-Trenaunay-Weber syndrome presented with gastrointestinal bleeding in Pakistan. Other reported cases of Kippel-Trenaunay-Weber syndrome from Sargodha, Multan and Bhawalpur were reported without gastrointestinal haemorrhage[8-10].

The most common bleeding sites in the gastrointestinal system are the distal colon and rectum. Jejunal haemangiomas and esophageal varices as bleeding sources caused by prehepatic portal hypertension were reported in the literature. Gastrointestinal haemorrhage usually begins in the first decade of life and tends to be intermittent.[8] However, the progressive nature of Kippel-Trenaunay-Weber syndrome warrants that physicians consider invasive surgical operation or angiographic intervention such as embolization of the bleeding vessel during the ongoing follow-up when there is a patient with transfusion-dependent anemia, life-threatening bleeding episodes, and/or poor quality of life due to severe anemia. Endoscopic therapy is usually preferred for localized lesions or postoperative residual disease.

One of the largest published series of Kippel-Trenaunay-Weber syndrome patients, haematochezia was reported in only six of 588 patients, although a few other cases may have gone unnoticed.[11] Differential diagnosis includes Parkes Weber syndrome, Proteus syndrome and the Maffuci syndrome. In Parkes Weber syndrome, limb hypertrophy is caused by multiple arteriovenous fistulae and hence a continuous brui may be present.

Conservative management and iron supplements may be sufficient in those patients who present with occasional non-significant and non-debilitating bleedings. However, long-term treatment mostly requires surgical resection comprising proctocolectomy in the case of life-threatening colonic bleeding and coloanal anastomosis with preservation of anal function, especially in younger patients, or abdomino - perineal resection in the case of rectal bleeding due to the diffuse and sometimes progressive disease process, anatomy and determining the disease extent vascular embolization can be considered if a distinct bleeding site is encountered. Endoscopic photocoagulation using argon laser is sometimes employed for the management of localized lesions or ablation of postoperative residual disease.

CONCLUSION

This report describes Klippel-Trénaunay syndrome as a rare cause of recurrent lower gastrointestinal bleeding. Apart from occasional hematochezia and mild anemia our patient was asymptomatic. In view of the distal 40 cm colonic involvement described ,bleeding will most likely only subside following surgical resection of the colon if there is severe hematochazia unresponsive to conservative management.in this case only oral iron supplementation was administered so far. We suggest regular follow up of these patients for the observation of complication and specific management.

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