Diffuse Cavernous Colo-Rectal, Visceral and Cutaneous Hemangiomas in Patient With Klippel-Trenaunay Syndrome: A Challenging Case For Management

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Diffuse Cavernous Colo-Rectal, Visceral and Cutaneous Hemangiomas in Patient With Klippel-Trenaunay Syndrome: A Challenging Case For Management

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Abstract

Background: Klippel-Trenaunay Syndrome (KTS) consists of a triad of cutaneous capillary hemangioma (Port wine stain), limb hypertrophy and venous varicosities. Gastrointestinal and genitourinary tracts vascular malformations have been reported as an association with KTS and can be a significant source of morbidity as rectal and bladder hemorrhage. Involvement of the gastrointestinal tract in KTS occurs in approximately 20% of patients with visceral involvement who mostly present with painless bleeding. Gastrointestinal involvement usually affects the distal colon and rectum.

Case description: In this article we report a 23 years old male patient with KTS associated with diffuse cavernous colo-rectal and visceral hemangiomas. Recurrent bleeding per rectum was the main complaint of the patient from the diffuse colo-rectal hemangiomas. In spite of applying different modalities for treatment starting from the age of 11, recurrence of symptoms occurred.

Challenge: Surgical intervention is deemed to be the last resort to solve the problem radically, but the selection of the type of surgery to be performed was difficult. While sphincter saving options as ultra-low anterior resection seem to be an optimal choice as they avoid permanent stomas and preserve continence, yet they carry a higher risk of anastomotic complications and eventual recurrence of bleeding. On the other hand, more radical surgery as abdominoperineal resection would definitely end the patient’s suffering and completely solve the problem, but at the expense of performing a permanent colostomy. Taking the decision as regards the type of operative intervention in this young patient sounds like a true challenge

KEYWORDS: Colo-rectal, hemangioma, Klippel, Trenaunay
Introduction
Diffuse cavernous hemangioma of the colon and rectum is a congenital vascular neoplasm or a hamartoma composed of dilated tortuous blood vessels in excess amount and haphazard distribution. [1] It is considered a rare disorder as only 350 cases approximately were reported all over the world since Phillips reported the first case in 1839. [2]

Colo-rectal hemangiomas are usually diagnosed in children and young adults. [3] The commonest location of these hemangiomas in the gastrointestinal tract is at the rectosigmoid region. [4] Less frequently they involve other regions of the gastrointestinal tract such as the small intestine and the stomach. [5] Also they may affect extraintestinal areas such as the liver, spleen, and bladder. [6] Many syndromes comprise gastrointestinal hemangiomas as a part of it such as Klippel-Trenaunay syndrome or Bean’s syndrome. [7]

Colo-rectal hemangiomas clinically manifest at early stage of the disease, the usual presentation is painless bleeding per rectum which might be acute, recurrent or chronic bleeding. The rectal bleeding could be in the form of melena or hematochezia. [8] Owing to the chronic or intermittent nature of this bleeding, anemia is a common finding in patients with colo-rectal hemangiomas. [9] The bleeding in some cases can be severe enough to be life threatening or to cause serious morbidities as consumptive coagulopathy, intestinal obstruction and perforation. [7]

Diagnosis of colo-rectal hemangiomas require a high level of suspicion since the presenting symptoms are non-specific and usually mistaken for hemorrhoidal bleeding or portal hypertension. [7] The delay time between the first clinical presentation and final diagnosis can reach up to 50 years. [10]

The diagnosis of cavernous colo-rectal hemangioma is often established at adulthood. Barium enema, colonoscopy and selective inferior mesenteric angiography are the diagnostic modalities used in reaching the final diagnosis. Computed tomography (CT) or better the magnetic resonance imaging (MRI) are mandatory to confirm the diagnosis and to evaluate the extent of the hemangioma, as they provide data about the dimensions of the lesion and whether the adjacent structures are involved or not. [4]

Complete surgical excision of the lesion is the primary mode of treatment as with conservative proctectomy with colo-anal anastomosis for isolated rectal hemangiomas. On the other hand, other lines of treatment have been suggested such as the injection sclerotherapy, argon laser fulguration, cryotherapy and colonoscopic banding.

In this report we are presenting a challenging case of colo-rectal hemangioma in a patient with Klippel-Trenaunay syndrome, which failed to resolve despite trying both, the surgical and the injection sclerotherapy options, thus making the management of such case a surgical dilemma and a true challenge.

Case Description
A 23-year-old male was admitted to our colorectal surgery unit with a complaint of recurrent bleeding per rectum and difficulty of defecation for a duration of one year.

**History of the case**

Going through the patient's medical history, he stated that this condition started since birth by a congenital vascular lesion in his right leg and ankle, with hypertrophy of the right lower limb and obvious varicosities in his right leg. This triad of vascular hemangioma, limb hypertrophy and varicosity highly suggested Klippel-Trenaunay syndrome. Color Doppler sonography was done on his right lower limb and varicose veins with incompetence of the sapheno-femoral junction were detected, however no arteriovenous malformations were found. The vascular lesion increased progressively in size, requiring surgical excision that was attempted three times.

At the age of eleven, he complained of bleeding per rectum that was painless, intermittent and associated with a mass prolapsing from the anus during straining. The recurrent bleeding was complicated by anemia manifesting as easy fatigue and drowsiness, necessitating transfusion of two units of fresh blood.

**First intervention**

He was admitted then to general surgery department and was submitted to a series of investigations including colonoscopy which revealed multiple cavernous hemangiomas in the descending and sigmoid colon with dilated tortuous rectal vein. Abdominal ultrasonography revealed focal lesions in the spleen and the liver assumed to be cavernous hemangiomas.

The decision to perform curative surgery for the colonic lesions was made and left hemicolectomy was performed. Following surgery the patient reported improvement of his symptoms and cessation of the rectal bleeding, yet he was still feeling a mass protruding from the anus while straining.

**Second intervention**

At the age of 16 years he complained again of recurrent fresh bleeding per rectum and mass prolapsing from the anus. The bleeding was mild and intermittent in nature. He was admitted to our colorectal surgery unit and examined. Vital signs were within the normal range. Abdomen was soft and non tender with evidence of scar of the previous hemicolecotomy. A vascular hemangioma was noted on his right lower limb with evident scars of previous operative interventions (Figure 1), hypertrophy and venous varicosities were also noted during examination of the right lower limb (Figure 2). During rectal examination, a 10 x 15 cm reddish pink mass was protruding from the anus with multiple dilated veins overlaying it. No other anal or perineal lesions were detected.

Upon admission, colonoscopy was done and revealed multiple dilated tortuous veins in the lower rectum with cavernous hemangiomas at the upper rectum (Figures 3, 4), the colorectal anastomosis was patent with no stricture noted. Beyond the anastomosis the rest of the colon was free of any vascular lesions or malformations.

Subsequently, anorectal examination under anesthesia (EUA) was performed. Prolapsing rectal mucosa with multiple dilated vascular lesions was found.
intraoperatively. Injection sclerotherapy with 10 ml of ethanolamine oleate was performed through the muco-cutaneous junction and a pack of gauze was inserted.

The patient was observed for one week postoperatively and no recurrent rectal bleeding or prolapsing rectal mass were observed. The patient was discharged on antibiotics and doxium as decongestant and mild analgesic.

**Third admission**

Seven years after the injection sclerotherapy session, the patient came to our outpatient clinic complaining of recurrent bleeding per rectum and difficulty of defecation for a duration of one year. He was re-admitted and a new colonoscopy was performed. Cavernous hemangiomas of the upper two thirds of the rectum were evident with few scattered dilated tortuous veins at the lower third of the rectum. No biopsy was taken due to the high risk of hemorrhage caused by manipulation of these vascular lesions.

CT angiography was ordered and it revealed a rectal hemangioma supplied by the terminal branches of the inferior mesenteric artery with contributing branches of both internal iliac arteries (Figure 5). Post-contrast CT abdomen and pelvis showed several scattered hemangiomas at segment VIII of the liver and mildly enlarged spleen with multiple focal lesions suggestive of splenic hemangiomas (Figure 6). A massive circumferential thickening of the upper rectum forming a mass-like lesion was noticed, with multiple foci of calcifications (Figure 7). The rectal lesion appeared to displace the bladder and small bowel loops around it, with surrounding perirectal haziness and enlarged perirectal and internal iliac lymph nodes. Non-contrast CT of the brain was done to exclude and cerebral vascular malformations and was free of any angiomatous malformations.

Owing to the diffuse nature of the rectal hemangiomas found in colonoscopy, further injection sclerotherapy or other conservative management were abandoned. Angiographic embolization of the lesion also was excluded, due to the circumferential nature of the lesion and the extensive feeding from inferior mesenteric and internal iliac arteries. Surgical intervention was deemed to be the last resort to solve the problem radically.

The selection of type of surgery to be performed was difficult. While sphincter saving options as ultra-low anterior resection seem to be an optimal choice as they avoid permanent stomas and preserve continence, yet they carry a higher risk of anastomotic complications and eventual recurrence of bleeding. On the other hand, more radical surgery as abdominoperineal resection would definitely end the patient's suffering and completely solve the problem, but at the expense of performing a permanent colostomy. Additionally, there are the grave complications of abdominoperineal resection, especially sexual dysfunction in a young male patient. Making the decision as regards the type of operative intervention in this young patient sounds like a true challenge.

Based on the patient’s history and persistent symptoms, the decision was made to do abdominoperineal resection as a radical cure for his suffering. However, the patient absolutely refused any further operative intervention, particularly abdominoperineal resection which would compromise the quality and style of his life. The patient chose to continue on conservative treatment in the form of venotonic as diosmin 500 mg...
combined with vitamin C tablet. One tablet was given three times per day for one week then twice daily for the next three weeks. Also hemostatic agents were prescribed as tranexamic acid 500 mg tablet. One tablet was given twice daily for one week. Vitamin K1 (phytonadione) 5 mg chewable tablet, one tablet was given twice per day for two weeks. In each subsequent follow up visit, the patient reported cessation of rectal bleeding with intermittent anal pain of mild intensity. So far, the patient remained free of rectal bleeding for a period of six months of regular follow up.

Discussion and Review of Literature
Hemangiomas are benign tumors consisting of excessive amount of mature normal structured blood vessels which are lined by a single layer of endothelium. They are one of the rarest tumors of the gastrointestinal tract. [11] Gentry et al identified only 96 cases of colo-rectal hemangiomas out of the 255 cases of gastrointestinal hemangiomas they studied.[12] About half of colo-rectal hemangiomas are located in the rectum. [13]

As regards age and sex distribution, colo-rectal hemangiomas have an equal sex distribution, which is not the case with small intestinal hemangiomas that occur in males more than twice they occur in females. [12] As a congenital disorder, hemangiomas tend to be symptomatic from childhood, but the diagnosis is usually not made until adulthood. [14] The patient in our study indeed was symptomatic at 11 years, yet he was diagnosed early by colonoscopy, which emphasize the importance of endoscopy in investigating cases of gastrointestinal bleeding whether upper or lower bleeding.

Colo-rectal hemangiomas can be solitary or multiple. They may be isolated lesions or associated with other synchronous lesions of the stomach and small intestine. Also, they can be a part of a systematic involvement of the spleen, kidney, brain, and skin. [15]

The patient in our report had a well-defined cutaneous hemangioma in his right lower limb, coping with what Gascoyen described in 1860 about the association of cutaneous lesions with GI hemangiomas. [16] Several syndromes have been described correlating cutaneous and gastrointestinal hemangiomas, such as blue rubber bleb nevus syndrome, Klippel-Trenaunay-Weber syndrome and Peutz-Jeghers syndrome. [17]

Kaijser classified gastrointestinal hemangiomas into four main categories which are: capillary hemangioma, cavernous hemangioma (localized, diffuse infiltrating and multiple phlebectasia), mixed hemangioma and hemangiomatosis. [18] The patient in our report had the localized polypoid subtype of cavernous hemangioma.

Capillary hemangiomas constitute 5 to 10% of benign vascular tumors of the gastrointestinal tract, being more common in the small intestine, appendix, and perianal skin. Usually they are solitary lesions that lack a true capsule. [19] Cavernous hemangiomas represent almost one third of benign vascular tumors of the gastrointestinal tract. They consist of larger blood vessels with a predilection for forming blood-filled sinuses. [20] Mixed hemangiomas share features of both capillary and cavernous hemangiomas and constitute 5 to 10% of benign intestinal vascular tumors. Disseminated intestinal hemangiomatosis refers to the presence of more than 50 intestinal hemangiomas spread throughout the GI tract. [13]
Colorectal hemangiomas are congenital in nature, arising from embryonic sequestrations of mesodermal tissue. They enlarge by the proliferation of their endothelial cells. The different histological types reflect anomalies occurring at different stages of stem cell development, with capillary hemangiomas at the initial stage and cavernous at the second stage. [21]

Twenty percent of colo-rectal hemangiomas are asymptomatic. The hallmark of clinical presentation of the symptomatic lesions is intraluminal hemorrhage. [22] Up to 90% of cases present with recurrent painless rectal bleeding that initiates in childhood and become more severe progressively with age [23] leading to iron deficiency anemia in more than half of the patients. [24]

Bleeding is more severe and frequent with the cavernous type more than the capillary type because of their greater size and larger blood volume. Also rectal lesions are more frequently symptomatic than proximal colonic lesions due to more trauma to the rectal mucosa by the hard fecal matter. In general, the cause of bleeding per rectum is the erosion of the wall of abnormal vessels through the mucosa into the intestinal lumen. [18]

Owing to their non-specific symptoms, the diagnosis of colo-rectal hemangiomas is often difficult and many cases are misdiagnosed as hemorrhoidal bleeding. A triad of intermittent rectal bleeding, multiple ectopic phleboliths and cutaneous lesions should raise suspicion of hemangioma as the cause of gastrointestinal hemorrhage. [21]

Diagnosis is made using several imaging modalities as barium contrast studies. Characteristic features of hemangioma on barium enema are: presence of a luminal filling defect, narrowing of the distal rectosigmoid and widening of the prerectal space as a mass effect of the lesion. [25]

CT scan of the abdomen and pelvis is not only useful in detecting the tumor in the form of heterogeneous thickening of the colonic wall, it also defines the extent of the lesion and excludes the presence of invasion into surrounding structures. [26] Superior to CT scan, MRI provides detailed anatomic images and demonstrates blood flow anomalies without the need for contrast medium. [27]

Selective mesenteric angiography has several advantages as its ability to detect hemangioma even in the absence of active bleeding and detection of synchronous lesions. The vascular lesions take the form of venous pooling or most commonly in the rectosigmoid region. [28]

Colonoscopy is essential in diagnosing patients with colorectal hemangioma. Hemangiomas appear as submucosal projections, ranging from a deep blue to a dull red colored. [13] The overlying mucosa is edematous, suggesting chronic inflammation and there may be several pinpoint bleeding areas which may give a false impression of inflammatory bowel disease. [29]

As regards treatment of colo-rectal hemangiomas, surgical resection remains the mainstay of treatment. Non-operative therapy so far is limited in most cases. Medical treatment include steroids especially in infantile hemangiomas and Interferon-a2a.
Radiation therapy is used successfully in controlling bleeding from rectal lesions but is not used for colonic or intestinal lesions because of the toxicity to the adjacent organs. [30] Endoscopy can play an important role in treating colo-rectal hemangiomas. Polypoid lesions with a narrow base can be removed by snare polypectomy with electrocautery. [31]

Operation for patients with rectosigmoid lesions includes limited colectomy, low anterior resection, or abdominoperineal resection with terminal colostomy. Abdominoperineal resection was the procedure of choice for the resection of hemangiomas involving the distal rectum until 1976, since then, sphincter preservation using rectal mucosectomy and pull-through coloanal anastomosis has become the commonly used procedure in young patients with benign disease. [29]

There are some issues concerning the patient in this report, such as the diffuse nature of hemangiomas, the associated hepatic, splenic and cutaneous hemangiomas, the previous therapies provided (surgery and sclerotherapy) and difficulty in choosing the type of operative intervention to treat the patient. All of these issues make this case a challenge for the colo-rectal surgeon.

Two questions should be answered before going ahead and performing the surgical resection intended. The visceral hemangiomas carry a serious risk for intraperitoneal hemorrhage which can be life threatening. Should they be given the priority in management, and if so which line of treatment should be most appropriate? The second question concerns the type of intervention that should be made regarding the rectal lesions. A sphincter saving option carries a higher risk of recurrent bleeding and anastomotic problems compared to a radical abdominoperineal resection which terminates the rectal bleeding problem but leaves a young patient with an end colostomy for a benign disease besides the sexual dysfunction that might occur with this kind of operations. Which is the preferred surgical approach?

References

**Figure 1:** Cutaneous lesion and scars of previous surgeries in the right lower limb
Figure 2: Evident hypertrophy and varicosities of the right lower limb
Figures 3 and 4: Colonoscopic appearance of rectal hemangioma
Figure 5: CT mesenteric angiography demonstrating branches feeding the rectal hemangioma
Figure 6: Hepatic and splenic hemangioma
**Figure 7:** Post-contrast CT of the pelvis demonstrating circumferential thickening of the rectum with multiple foci of calcification (rectal hemangioma)