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Hemangiomas and vascular malformations of the GI tract

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ABSTRACT

Hemangiomas and vascular malformations of the GI tract are a rare entity and present as overt or occult bleeding. Presenting as a singular cavernous hemangioma or malformation, which is often located in the rectosigmoid region, they can be distributed throughout the intestinal digestive system. Despite characteristic radiographic features such as radiolucent phleboliths on plain film imaging and a purplish nodule on endoscopy, misdiagnosis is common. There is potential for local invasion, therefore adjunctive imaging such as CT and MRI are part of the suggested workup. Endorectal ultrasound with Doppler has also been found to be useful in some instances. With an emphasis on sphincter preservation, surgical resection is the mainstay of treatment, though in instances where an extensive resection is not feasible, non-surgical endoscopic treatment with banding and sclerotherapy has been reported with success.

Epidemiology

Hemangiomas and vascular malformations of the GI tract, first documented in 1839, are an infrequently encountered entity.[1,2] Potentially occurring anywhere in the gastrointestinal tract, the small bowel is the most frequent site of hemangiomas and malformations, though they account for 10% of all small bowel tumors.[3] With only 200 cases documented from 1931 to 1974, colonic and anorectal hemangiomas and malformations are even more rare entities.[4,5] A historical review in 1949 of GI hemangiomas and malformations identified 38% of hemangiomas as colorectal in origin, whereas another review classified 50% of colorectal lesions located in the rectum.[2,6]

To date, there have been over 130 reports of rectal hemangiomas/malformations.

The age of a definitive diagnosis of GI hemangiomas and malformations range from 2 months to 79 years old due to the fact that many are misdiagnosed.[2,7,8] There is a slight female preponderance with a male:female ratio of 1:2.5, though colonic hemangiomas and malformations have an equal male:female ratio.[2] Looking at the subclassification of vascular malformations known as blue rubber bleb nevus syndrome, the largest operative single-institution series ($n = 10$) identified a mean patient age of 16 years, with a range of 2–36 years.[9] The initial bleeding symptoms were present early (mean = 5 years).

Histology, pathology, and classification

Complicated by the confusing nomenclature, the term "hemangioma" is often misused. Modifiers and adjectives such as "strawberry," "cavernous," and "capillary" are added to the term "hemangioma," but are not hemangiomas in the strictest definition.[10,11]

In 1982, Mulliken and Glowacki classified vascular lesions in a consistent and meaningful scheme based on the histology and endothelial cell turnover.[12,13] Normal endothelial conditions are correctly termed "vascular malformations," classified by their dominant abnormality (venous malformation, arteriovenous malformation, lymphatic malformation, lymphatic-venous malformation, and capillary malformation).[11,14] High endothelial cell turnover entities are accurately termed hemangiomas (rapidly involuting congenital hemangioma, infantile hemangioma, noninvoluting congenital hemangioma, Kaposiform hemangioendothelioma, and tufted angioma). Present at birth, the majority of these lesions undergo a spontaneous involution. Steroid administration can accelerate the involution process. The International Society for the Study of Vascular Anomalies approved this classification system in 1996 (Table 1).

Until this point, this article used the term "hemangioma" due to the audience's familiarity with the term. Though hemangiomas do occur in the GI tract, the predominant entities are vascular malformations, including those termed as "cavernous hemangiomas." In precise terms, this is a misnomer, and the term "cavernous vascular malformation" or "cavernous malformation" is more accurate and will be used.

An embryologic error in morphogenesis is how vascular malformations result.[14] Lacking smooth muscle, mature endothelial

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Table 1
Vascular malformations versus hemangiomas.[11]

	Vascular malformation	Hemangiomas
Histology	Normal endothelial cell turnover.	High endothelial cell turnover.
Presence at birth	Present (not always apparent).	Usually absent.
Clincal	Grows in proportion with person.	Apparent 6–8 weeks after birth. Proliferative phase for 1–2 years then spontaneous involution.
Diagnosis	Imaging (MRI, CT, US, and angiography)	Clinical history and appearance.
Treatement	Depending on site, size symptoms, etc. from conservative-only to laser for capillary malformations, sclerotherapy with or without excision, or surgery alone.	Observation: If not fully involuted, minor surgical correction. If large or in anatomically sensitive area, steroids or interferon gamma or surgical correction.

channels allow expansion over time from hydrostatic means and not proliferative expansion as hemangiomas do. A classification of intestinal malformations was devised based on the histological abnormality (Table 2).

The capillary subtype of malformations is located in the small bowel, appendix, and perianal skin.[20] They lack a true capsule, are usually singular, and are well circumscribed. With accompanied edema and inflammation, half of them can have associated mucosal ulceration. The proliferation of capillaries with thin-walled spaces lined by endothelial cells is the histological hallmark.

The cavernous subtype composes 80% of rectosigmoid malformations.[16–18,21,22] Cavernous malformations are large spaces lined by single or multiple layers of endothelial cells, as opposed to capillary malformations (Fig. 1). Often polypoid, the localized variety of cavernous malformations can be symptomatic. Reported up to 30 cm in length, the diffuse variety of cavernous malformations can be multiple. With the rectum involved in 70% of instances, these entities have the potential for local invasion to adjacent structures and can be circumferential.[2,23]

Ranging from solitary lesions to clusters, the gross appearance of GI vascular malformations overwhelmingly present as intraluminal lesions, though diffuse cavernous malformations can infiltrate the submucosa and beyond and can extend to adjacent structures.

GI vascular malformations can be a feature of associated syndromes with characteristic organ involvement. Simultaneous vascular malformations in the spinal cord, brain, and skin as well as other clinical traits such as bone and limb hypertrophy, arteriovenous fistulas, and varicose veins have been documented (Table 3).[8,16,17,20,24] Only 1.8% of cutaneous hemangiomas and vascular malformations have GI vascular malformations present, despite cutaneous involvement being a frequent association.[25] No evidence exists of predisposed inherited patterns when analyzing monozygotic and dyzygotic twins.[26] However, this counters a case report describing 6 families that demonstrated an autosomal morphology with incomplete penetrance.[27]

Pathophysiology

The key differentiation between hemangiomas and vascular malformations in the original classification by Mulliken and Glowacki is due to the smooth muscle proliferation.[12,13] Related to the defect intrinsic to the endothelial cells and secretion of growth factors, the abnormality arises in the embryological mesoderm.[28] The 3:1 female to male ratio is thought due to hormonal influence.[2] The different histological subtypes are thought due to the developmental stage of when the angiogenic abnormality occurred in the stem cell cycle, with capillary malformations developing from an earlier defect than cavernous malformations.[2,8,16]

Table 2
Intestinal hemangioma classification.[15–19]

Capillary
Cavernous
Localized (polypoid or non-polypoid)
Diffuse infiltrating (expansive)
Mixed
Hemangiomatosis

Alteration of blood flow from the abnormal endothelium can sequester platelet and clotting factors and initiate coagulation.[29] Kasabach–Merritt syndrome can develop in rapidly expanding malformations, where a DIC-like picture can develop with consumption of fibrinogen and factors V and VIII, uncontrolled bleeding, and a 35% mortality rate.[30] Calcification and phlebolith development from the constant sequestration of flow can be seen in 50% of cases.[31] Erosion of the malformation into the bowel lumen and subsequent bleeding and also fragmentation of reticulocytes by the thrombus can lead to anemia. Local or segmental bowel ischemia can result from the sequestration and coagulation.[16] Case reports describe invasion the sacrum, bladder, and uterus of the hemangioma/malformation into surrounding structures, especially when the abnormality originates in the rectosigmoid region. Malignancy is rarely encountered, despite the ability for local invasion.[8,32,33]

Clinical presentation

History

Misdiagnosis of the hemangioma and malformation is a common theme. Overall, 80% of patients underwent one prior inappropriate surgical procedure.[34–36] A cases series identified that 4 out of 5 patients with an eventual diagnosis of a vascular malformations first underwent a hemorrhoidectomy.[37] GI bleeding was misdiagnosed as hemorrhoids and ulcerative colitis in another series evaluating rectosigmoid cavernous vascular malformations.[34,35] Evaluating 47 patients, another series estimated a delay in diagnosis of 16 years.[38] With similar conclusions, a separate review estimated that the correct age of diagnosis was at 19 years, with 51% of the patients undergoing either an inappropriate or ineffective operation.[36]

Intraluminal bleeding is present in a majority of cases, and an estimated 80% of patients exhibit symptoms.[39–41] Recurrent painless bleeding is seen in up to 90% of cases, with half of the patients showing chronic iron-deficient anemia. With episodes worsening over time, the first presentation is often in childhood.[6] There is potential for intraperitoneal or retroperitoneal bleeding when transmural malformations exist. Hemothorax or hemopericardium can present when there are non-GI malformations. Mucosal trauma causes erosion and eventual bleeding with the increased bleeding occurring in larger and more distal subtypes.

Though infrequent, obstruction is possible.[42] Circumferential masses can cause luminal obstruction, and polypoid lesions can act