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(CASE REPORT)

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Sporadic sigmoid colonic angiomyolipoma presenting as a pedunculated polyp: A case report and brief literature review

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Abstract

Angiomyolipoma (AML) is a benign tumor that is commonly found in the kidney. It is composed of blood vessels, smooth muscle, and fat. These mesenchymal tumors are very rarely found in the digestive tract, particularly within the sigmoid colon. We present the case of a 38-year-old female who presented with a four-month history of intermittent left lower quadrant abdominal discomfort, altered bowel habits, and occasional hematochezia. Colonoscopic evaluation revealed a 2.8 cm pedunculated polyp in the sigmoid colon with characteristics suggestive of a submucosal lesion.

Following multidisciplinary discussion, the patient underwent segmental resection of the sigmoid colon. Histopathological examination demonstrated the characteristic composition of AML. Immunohistochemistry (IHC) studies were positive for Melan-A, HMB-45, and smooth muscle actin. The Ki-67 proliferation index was low, and no atypia or necrosis was identified, confirming the diagnosis of AML.

This case highlights the significance of considering AML in the differential diagnosis of submucosal colonic polyps. Its diagnosis relies on histopathological and IHC evaluation, and complete resection remains both diagnostic and therapeutic.

Keywords: Angiomyolipoma; Perivascular epithelioid cell tumors, Renal; Extrarenal; TSC1/TSC2–mTOR signaling pathway

1. Introduction

AML is a rare mesenchymal neoplasm composed of varying proportions of mature adipose tissue, dysmorphic blood vessels, and smooth muscle cells. It belongs to the family of perivascular epithelioid cell tumors (PEComas), a distinctive group of lesions characterized by melanocytic and smooth muscle marker expression. [1]

AML and PEComa are not exactly the same, but they belong to the same family of tumors. PEComa is the broad umbrella term for a specific type of mesenchymal tumor, and AML as the most common and well-known member of that family. [2] The main family members of PEComa include: angiomyolipoma (AML), lymphangioleiomyomatosis (LAM), clear cell sugar tumor (CCST), and other soft tissue PEComas. [3]

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AML most commonly arises in the kidney, where it may occur sporadically or in association with tuberous sclerosis complex. In contrast, extrarenal AML is distinctly uncommon, and gastrointestinal involvement is exceptionally rare. Within the gastrointestinal tract, colonic AML is an exceedingly rare entity, described mainly in isolated case reports. [4,5]

Because of its rarity and nonspecific presentation, this lesion can be mistaken for more common colorectal polyps or other mesenchymal tumors, such as lipoma, leiomyoma, and gastrointestinal stromal tumors. [1,5]

Patients may present with abdominal pain, gastrointestinal bleeding, anemia, or altered bowel habits, although the lesion may also be found incidentally during a routine colonoscopy. [6] Imaging and endoscopic findings are not diagnostic, and definitive diagnosis usually requires complete excision followed by histopathologic and IHC evaluation. Recognizing this lesion is clinically important because most conventional AMLs are benign and can be treated with local excision, whereas epithelioid variants may behave more aggressively. (See table 1)

We report this case to highlight the diagnostic challenges, clinicopathologic features, and successful management of an incidental sporadic AML of the sigmoid colon presenting as a pedunculated colonic polyp.

2. Case Presentation

2.1. Clinical Presentation

A 38-year-old female presented to the outpatient gastroenterology clinic with a four-month history of intermittent left lower quadrant abdominal discomfort associated with mild changes in bowel habits, specifically alternating episodes of constipation and loose stools. She reported occasional passage of bright red blood per rectum, which she initially attributed to hemorrhoids. The patient denied any significant weight loss, nocturnal symptoms, or fever. Her appetite remained intact throughout the symptomatic period. Her past medical history was unremarkable.

On physical examination, the patient appeared well-nourished and in no acute distress. Vital signs were within normal limits. Abdominal examination revealed mild tenderness to deep palpation in the left iliac fossa without guarding or rigidity. No palpable mass was appreciated, and bowel sounds were normal. Digital rectal examination was unremarkable.

2.2. Early Investigations

Routine laboratory investigations, including complete blood count, comprehensive metabolic panel, and coagulation profile, were within normal limits. Serum carcinoembryonic antigen (CEA) was within the reference range. The stool occult blood test returned positive, which, in conjunction with the patient's lower gastrointestinal symptoms, prompted referral for colonoscopy evaluation.

Colonoscopy was performed following standard bowel preparation. The examination extended to the cecum and revealed excellent mucosal visualization throughout the colon. At 18 cm from the anal verge, within the sigmoid colon, a well-defined pedunculated polyp was identified. The lesion measured approximately 2.8 cm in its greatest dimension and was noted to have a broad base with a distinct, wide stalk. The overlying mucosa appeared intact and smooth, with a characteristic yellowish hue, a feature that raised immediate consideration for a submucosal process.

Endoscopic assessment demonstrated that the lesion was submucosal in origin, with the overlying mucosa freely mobile over the mass on probing, the so-called "pillow sign" or cushion sign, consistent with a submucosal mesenchymal lesion. No additional polyps or mucosal abnormalities were identified in the remainder of the colon or the terminal ileum. Differential diagnosis included benign mesenchymal tumors (specifically lipomas and AMLs), vascular lesions, and malignant neoplasms that can be presented as large polyps. Of note, there was no personal or family history of tuberous sclerosis complex (TSC), no known syndromic diagnosis, and no prior history of colonic polyps or colorectal malignancy. She had no history of renal angiomyolipomas, pulmonary Lymphangioleiomyomatosis, or dermatologic stigmata associated with TSC.

2.3. Multidisciplinary Tumor Board Discussion, Management, Pathological Findings, and Outcome

At the multidisciplinary gastrointestinal tumor board, treatment options were discussed, and endoscopic resection was preferred over surgical segmental resection. The patient was informed that the most likely diagnosis was a benign colonic polyp. However, the patient chose segmental resection due to concerns about potential excessive bleeding and

possible unclear pathology results. Consequently, a segmental resection of the sigmoid colon, which included the entire polyp, was performed.

The specimen was submitted for histopathological analysis. Grossly, the resected specimen showed a polypoid mass measuring 2.8 × 2.1 × 1.7 cm, with a smooth external surface and a pale yellowish-tan, lobulated cut surface on sectioning. Microscopic examination revealed a submucosal lesion composed of a characteristic triphasic admixture of mature adipocytes, thick-walled dysmorphic blood vessels, and bundles of smooth muscle cells arranged in varying proportions, the hallmark histological triad of AML. The overlying colonic mucosa was intact without evidence of dysplasia or malignancy. The smooth muscle cells demonstrated abundant eosinophilic cytoplasm and occasional cytoplasmic clearing. (Figure 1 A, B, C)

IHC studies confirmed the diagnosis: the spindle and epithelioid cells were diffusely positive for the melanocytic markers HMB-45 and Melan-A, and smooth muscle actin while staining negative for S-100 protein, cytokeratins, and CD34. The Ki-67 proliferation index was low (<2%), and no nuclear atypia, necrosis, or atypical mitoses were identified, arguing strongly against malignant features. These findings were diagnostic of a benign colonic AML belonging to the PEComa family.

Complete endoscopic resection served as both a definitive diagnostic and therapeutic intervention. Given the benign histomorphology with supportive IHC, low Ki-67, and absence of histological features of malignancy, no further oncological treatment was warranted. Long-term colonoscopy follow-up was recommended, given the paucity of natural history data for this exceedingly rare entity.

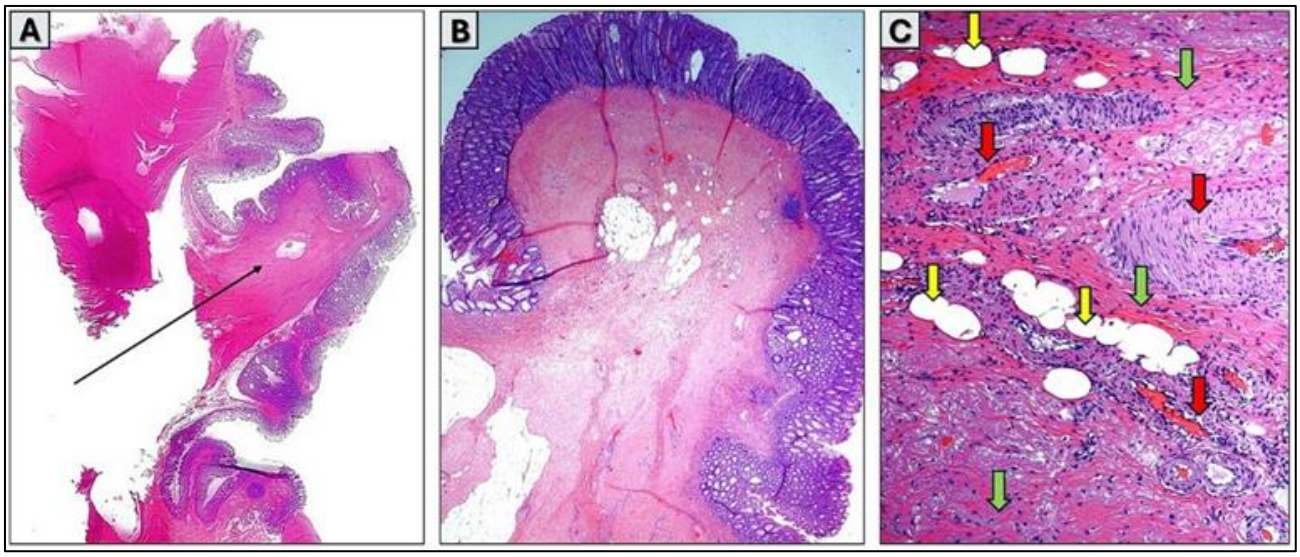

Figure 1 Microscopic findings of the excised colonic angiomyolipoma (AML)

- 1A: Full low-power view showing the entire 2.8 polypoid pedunculated mass, bisected, black arrow points to a broad base with a distinct wide stalk (H&E stain X10).
- 1B: Low power view showing the pedunculated polypoid angiomyolipoma mass with a broad stalk and smooth, intact overlying mucosa (H&E stain X20)
- 1C: High power view showing the three components of angiomyolipoma, thickened blood vessels (Red arrows), fatty tissue (Yellow arrows), and smooth muscles (Green arrows) (H&E stain X40).

3. Discussion

3.1. Background (History, epidemiology, risk factors, and WHO classification):

AML is currently classified as a member of the PEComa family, a unique neoplasm with dual melanocytic and smooth muscle differentiation. [7] AML was first diagnosed in kidney in the early 1900s. [8] However, extrarenal forms involving the liver, retroperitoneum, uterus, skin, and, rarely, the gastrointestinal tract was identified in later reports. [1,3,5] Renal AML is the most prevalent subtype, comprising ~1% of surgically resected renal masses, whereas colonic AML is extremely rare, with the literature reporting only isolated case reports. [9] Table 1 summarizes the nomenclature

of renal and extrarenal PEComa. The nomenclature of renal and extrarenal PEComas unifies a broad spectrum of histological and IHC related mesenchymal neoplasms. Because these tumors frequently share an origin from perivascular epithelioid cells (PECs), they are classified based on anatomic location, histological composition, and whether they are pure or mixed neoplasms. [24]

Most lesions are benign, but epithelioid variants may have an aggressive potential. The current WHO classification classifies AML as PEComa, with risk determined by size, atypia, mitotic activity, necrosis, and infiltrative growth. Sigmoid colonic AML is a rare condition with diagnosis and therapy based only on published case reports. [7,10]

Table 1 Nomenclature of renal and extrarenal PEComa

IF Microscopic Appearance	Found in the Kidney (Renal)	Found in Soft Tissue / Organs (Extrarenal)
Triphasic (Fat + Muscle + Vessels)	Classic Renal AML	Extrarenal AML
Pure Epithelioid Cells (High cellularity)	Epithelioid AML (or Renal PEComa)	PEComa (NOS / Malignant PEComa)

[*References 6,7]

AML can occur both in sporadic setting and in TSC, resulting from mutations in TSC1 or TSC2. More frequently, syndromic cases are multifocal, bilateral, or detected at younger ages. [11] No known environmental or occupational risk factors are reported. [5] Gastrointestinal AML is most commonly a middle-aged condition with signs and symptoms of abdominal pain, bleeding, anemia, intussusception, and incidental endoscopic diagnosis. [12]

3.2. Pathogenesis, Pathophysiology

AML is best understood as part of the PEComa family. The triphasic architecture of AML explains both its benign polypoid appearance and its potential clinical effects: the adipocytic component could produce a yellow submucosal mass, the smooth-muscle component could form a firm stalk or nodule, and the abnormal vascular component could predispose to mucosal irritation, occult bleeding, or overt hematochezia when the lesion enlarged, prolapsed, or ulcerated. [12,13]

At the molecular level, many AMLs and PEComas appear to be driven by dysregulation of the TSC1/TSC2–mTOR signaling pathway. TSC1 and TSC2 (Tuberous sclerosis complex 1 & 2) encoded hamartin and tuberin, which normally suppress mTOR-mediated cell growth; loss or functional impairment of this complex permits increased downstream mTOR activity, protein synthesis, and cellular proliferation.[14] Importantly, mTOR pathway activation was demonstrated not only in TSC-associated angiomyolipomas but also in sporadic, non-TSC AMLs and extrarenal PEComas, supporting a shared pathogenetic mechanism even in the absence of syndromic features.[14,15] A smaller subset of PEComas showed alternative molecular events, including TFE3 rearrangement or overexpression, which may define biologically distinct tumors.[16,17]

The behavior of colonic AML remained difficult to predict because reported cases were scarce. Most classic triphasic colonic lesions behaved benignly after complete excision, particularly when they were small, circumscribed, mitotically inactive, and lacked necrosis or cytologic atypia. [12,18] In contrast, PEComas with high-risk features such as size greater than 5 cm, infiltrative growth, high nuclear grade, necrosis, vascular invasion, or increased mitotic activity were associated with uncertain or malignant potential. [19] In the present case, the intact mucosa, low Ki-67 index, absence of atypia, necrosis, and atypical mitoses, and complete excision supported a benign sporadic colonic angiomyolipoma rather than an aggressive PEComa.

3.3. Comparative Analysis of Our Case with Existing Literature. (Clinical, radiology, pathology, lab, diagnosis, management, and outcome)

The present case shared several features with previously reported colonic AMLs but also offered useful distinctions. Similar to the earliest sigmoid case described by Hikasa et al. and the large pedunculated sigmoid lesion reported by Sharara and Tawil, this tumor presented as a smooth, yellowish, polypoid submucosal lesion in the distal colon and showed the classic triphasic composition of fat, dysmorphic vessels, and smooth muscle. [12,13] Its size of 2.8 cm also fell within the commonly reported range for colonic AMLs, which have often been described as polypoid or pedunculated lesions measuring approximately 1–5 cm, with a predilection for the left colon and no clear association with TSC. [18]

However, this patient differed from much of the earlier literature because she was a young woman, whereas classic colonic AML had been reported predominantly in middle-aged or older men. [12,13,18] Her intermittent rectal bleeding and bowel habit changes resembled the symptomatic sigmoid case reported by Tabet Aoul et al., in which a 3-cm semi pedunculated sigmoid AML caused hematochezia and constipation. However, our patient lacked anemia and had a surgically resected pedunculated lesion. [18] Larger lesions, such as the 5.7-cm descending-colon AMLs described by Oishi et al. and Gandhi et al., required colectomy because of hemorrhage risk or intussusception. [20,21] In contrast to colonic PEComa reports showing HMB-45 positivity with atypia, necrosis, increased mitoses, or TFE3 expression, the present lesion retained benign morphology and a very low proliferative index. [16,22,23]

4. What Have We Learned from This Case?

This case highlights the importance of keeping a broad differential when evaluating colonic polyps, especially in younger female patients. Although most colorectal polyps are epithelial in origin, a yellow, submucosal-appearing lesion with a positive "pillow sign" should raise concern for a mesenchymal tumor, such as lipoma or, much rarer, AML. In this patient, mild lower GI symptoms and a positive stool occult blood test prompted colonoscopy, during which the lesion's submucosal characteristics guided further diagnostic testing.

One important takeaway from this case is that not all colonic polyps represent the typical adenomatous or malignant processes we often think of first. Rare benign lesions, such as AML, can present features that raise concern for more serious pathology. This emphasizes the role of tissue diagnosis, as histology and IHC were necessary to distinguish this lesion from other submucosal tumors.

The combination of abnormal thick-walled vessels and smooth muscle cells supported the diagnosis, while positive HMB-45 and Melan-A staining confirmed the diagnosis of AML. Reassuringly, the low Ki-67 index and absence of atypia or abnormal mitotic activity supported a benign course. For future practice, this case reinforces the need to recognize atypical polyp features, involve pathology early, and individualize management and surveillance when evidence is limited.

Abbreviations

- Angiomyolipoma (AML);
- Immunohistochemistry (IHC);
- Perivascular epithelioid cell tumors (PEComas);

5. Conclusion

This case illustrated that sporadic AML of the sigmoid colon, although exceedingly rare, could mimic a conventional pedunculated colonic polyp or lipomatous submucosal lesion during colonoscopy. Its recognition required careful correlation of endoscopic appearance, triphasic histology, and IHC, particularly because gastrointestinal PEComas may overlap clinically with lipoma, GIST, vascular lesions, and malignant mesenchymal tumors. The absence of TSC features, dysplasia, necrosis, atypical mitoses, and increased proliferative activity supported a benign lesion, yet the rarity of this entity justified clinical surveillance.

We present this case to expand the limited literature on colonic AML, especially in a young female patient with a pedunculated sigmoid lesion and intermittent rectal bleeding. For the medical community, the value of this report lies in reinforcing the importance of AML as a rare but important differential diagnosis for yellowish submucosal colonic polyps and in emphasizing that complete excision with histopathologic confirmation remains both diagnostic and therapeutic.

Compliance with ethical standards

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Disclosure of conflict of interest

All authors make the following declarations:

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Financial relationships: All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might be interested in the submitted work.

Statement of ethical approval

The present research work does not include any studies on animal or human subjects conducted by any of the authors. This study was conducted as a retrospective review of archival pathology material collected during routine clinical care. All data were fully de-identified prior to analysis.

Statement of informed consent

Informed consent for the publication of this case was obtained from the patient.

Data access statement

All relevant data are included in the paper.

Author contributions

All authors contributed equally to producing this manuscript.

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