



Angiomyolipoma of the Colon, Case Report of Uncommon Tumor and Review of the Literature

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Abstract

Angiomyolipomas (AMLs) are benign mesenchymal tumors of generally unknown origin, that consist of mature adipose tissue, smooth muscle fibers and blood vessels with thickened wall, most commonly seen arising in the kidney. Angiomyolipomas comprise about 1% of all renal masses. In contrast, extrarenal retroperitoneal AMLs represent extremely rare tumors with fewer than 100 reported cases. Colon angiomyolipomas is considered a rare site of this tumor. They are usually asymptomatic and present a unique diagnostic challenge since they can mimic other benign and malignant tumors, which must be differentiated. We report a case of a 58-year-old female with submucosal polypoid sigmoid lesions. Histopathological examination was diagnostic of colonic angiomyolipoma. We discuss differentiation of this entity from other tumors and we provide brief review of the literature

Keywords: Angiomyolipoma; Renal; Extrarenal; Tuberos sclerosis; Mesenchymal

Abbreviation

AMLs: Angiomyolipomas, **TSC:** tuberous sclerosis complex, **IHC:** Immunohistochemistry

Introduction

Angiomyolipoma (AML) is a mesenchymal tumor derived from perivascular epithelioid cells and consists of blood vessels, muscle cells, and fat tissue [1]. Histologically they are composed of thick-walled vessels, smooth muscle cells, and variable amounts of mature adipose tissue [2]. Angiomyolipomas are rare lesions, often arising in the kidney, and are part of a group of tumors with a diverse appearance and evidence of dual melanocytic and smooth muscle differentiation known as PEComas (tumors of perivascular epithelioid cell origin) [3]. Extrarenal angiomyolipoma is uncommon, but most commonly found to occur in the liver and less commonly in the spleen, lungs, retroperitoneum, bone, and ovaries [4-5]. In most cases, angiomyolipomas are benign in the clinical course, even those with multiple foci, a bizarre morphology, or local invasion [6]. Angiomyolipoma of the colon is a very uncommon extrarenal site. We report a case of a 58-year-old female with a sigmoid lesion diagnosed pathologically as angiomyolipoma of the colon and perform a brief review of the literature.

Case Presentation

A 58-year-old female presented with a complaint of blood tinged stool in addition to frequent abdominal pain. Patient reported history of controlled diabetes mellitus and mild controlled hypertension. No other significant history was reported. Colonoscopy was performed and a mid-sigmoid polypoid 2.5 x1 cm lesion was identified and appeared to be submucosal.

An enhanced computed tomography (CT) scan of the lower abdomen revealed a 2.5 × 1 cm well defined non enhancing polypoid submucosal soft tissue mid sigmoid colon mass. Imaging studies were inconclusive, and a tumor board multidisciplinary meeting recommended segmental resection of the sigmoid colon including the mass. The excised surgical specimen was 11.0 cm in length and 5.5 cm in circumference. Opening of the colon segment revealed a pink-tan pedunculated polypoid submucosal mass (2.5 x 1.5 cm) with a stalk of 0.5 cm in length (**Figure 1A**). The mass was 3.5 cm from the closest surgical margin. The colonic mucosa was otherwise unremarkable. Microscopic examination revealed an admixture of three components of tissue, mature adipose tissue, irregular thick-walled vessels, and interspersed areas of bland spindle smooth muscle cells. The thick vessels were the dominant component of the mass and were intermingled with the adipose cells and smooth muscles. There was no significant nuclear atypia, pleomorphism, or abnormal mitoses (**Figure 1B-C**). IHC studies were utilized for further evaluation. The tumor cells were positive for vimentin, desmin, SMA (**Figure 1D**) and scattered spindle cells were stained with HMB- 45 (**Figure 1E**). Rare cells stained with CD34 (**Figure 1F**) The tumor cells were negative for S100, and pancytokeratin.

The histomorphology and IHC profiled were diagnostic of angiomyolipoma of the colon with no evidence of atypia or malignancy. Patient was followed for three years with no evidence of recurrence after which she was lost to follow up.

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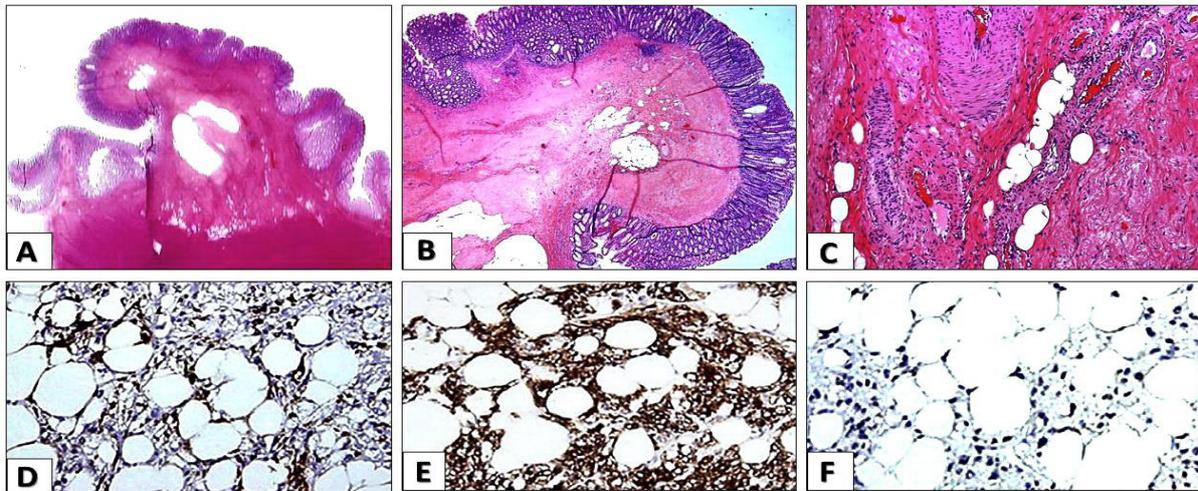


Figure 1 Pathological examination of the excised angiomyolipoma.

1A: Colon segment revealed a pedunculated polypoid submucosal mass, Low power view (H&E stain x10)

1B: Pedunculated polypoid submucosal mass, intermediate power view (H&E stain x20)

1C: Admixture of three components of tissue, mature adipose tissue, irregular thick-walled vessels, and interspersed areas of bland spindle smooth muscle cells (H&E stain x60)

1D: Tumor cells focally positive for HMB-45

1E: Tumor cells positive for SMA

1F: Rare cells positive for CD34

Discussion

Angiomyolipomas (AML) are unusual mesenchymal neoplasms composed of blood vessels, smooth muscle, and adipose cells [7]. These tumors have the potential for local invasion and distant metastases, but the involvement of regional lymph nodes by these tumors is uncommon [11]. It has also been reported that angiomyolipoma has some similarities with lymphangiomyomatosis (LAM) [12]. AMLs represent 0.3-3% of all renal tumors, with a female preponderance, due to hormonal influences [13]. Angiomyolipoma was first described by Morgan in 1951 [8]. Although angiomyolipoma is considered a benign tumor, a malignant epithelioid variant has been described with majority of cases metastasizing [9-10].

Renal angiomyolipoma's (RAML) is the most commonly reported type. There are 2 types of RAML, sporadic angiomyolipomas and tuberous sclerosis complex (TSC)-associated angiomyolipomas. TSC-associated angiomyolipoma's is an autosomal dominant disease and account for approximately 20% of cases [14]. Association of tuberous sclerosis with AML is not limited only to renal site but has been also reported in some Extrarenal AMLs [15].

TSC2 inactivation by mutation is a consistent and likely necessary genetic event in the pathogenesis of most angiomyolipoma [16]. TSC-associated AML develops at a younger age and tends to exhibit a much faster growth rate over time than sporadic AML [17]. Patients with TSC-associated AML develop multifocal and bilateral tumors, with a high rate of recurrence in comparison to sporadic AMLs [18]. About 80-90% of cases, renal or extrarenal, are sporadic, and these are most commonly found

in middle-aged women. Sporadic AML is usually asymptomatic and grows slowly. Active surveillance is generally conducted for patients with small (<4 cm) sporadic AMLs [19-20]. Diagnosis of AML can be challenging. Adding to the difficulty of a preoperative diagnosis, most extrarenal AMLs are asymptomatic and remain occult unless incidentally detected [6].

Most of these tumors are asymptomatic and detected incidentally. Larger tumors (>4 cm) can be symptomatic with flank pain and hematuria or following retroperitoneal hemorrhage from intra-tumoral vessels [21]. Angiomyolipomas larger than 5 cm and those containing an aneurysm pose a significant risk of rupture, which is a medical emergency, as it is potentially life-threatening. One population study found the cumulative risk of hemorrhage to be 10% in males and 20% in females [22]. The main causes of AML rupture are an increase in the circulating blood volume, an elevation of blood pressure, and an increase of pressure on the AML in association with uterine growth. A larger tumor size, younger patient's age and higher BMI value correlated with a higher risk of tumor hemorrhage [23-24].

Extrarenal AMLs, however, are more difficult to diagnose on imaging as they often lack fat densities [25]. The current diagnostic methods include ultrasound, Computed Tomography (CT) and Magnetic Resonance Imaging (MRI). Because of its fat component, the preferred diagnostic method for AML is CT [26]. On a CT scan with contrast agent, they show a prolonged enhancement pattern homogeneous with peripheral kidney tissue. Classic AML usually appears as a hyperechoic and homogeneous lesion in comparison to the normal renal parenchyma [27-28]. Classic AMLs appear hyperintense on T1- and T2-weighted images and hypointense on T1-weighted images after selective fat suppression on MRI [29].



Immunohistochemistry (IHC) studies reported that all of the AML of the colon reacted to SMA. Most renal AML react to HMB-45, but AML of the colon reacting to HMB-45 is rare. In our case only focal scattered cells were positive for HMB-45. The tumor cells reacted to desmin and HMB-45 in some reported cases [30]. Renal angiomyolipomas are commonly seen in patients with tuberous sclerosis. Despite their multiplicity, they rarely are prone to aneurysm formation or rupture when measuring less than 4 cm. Small lesions can, therefore, be closely monitored via imaging. AMLs larger than 4 cm have greater propensity for acute bleeding; thus, they are amenable to angiography and embolization, as opposed to traditional surgical therapeutic options even when asymptomatic [31]. According to the recommendations of the International Tuberous Sclerosis Complex Consensus Conference (ITSCCC) for TSC-associated AML, abdominal MRI or US should be performed to assess the progression of AML every 1–3 years throughout the lifetime of the patient [32]. Small asymptomatic tumors may be managed with active serial imaging surveillance; however, large asymptomatic AML should be removed to avoid spontaneous rupture due to the presence of abnormal elastin in the tumor [33]. Transarterial embolization is recommended as a first-line treatment for bleeding AML. The ITSCCC recommended mTOR inhibitors as a first-line treatment for AML of ≥ 3 cm in size, even when asymptomatic [32]. The major aims of treatment for TSC-associated AML are to maintain renal function and to prevent rupture and enlargement of the AML [34].

Angiomyolipomas are often benign mesenchymal lesions mostly occurring in the kidney. Extrarenal angiomyolipomas are uncommon, but most commonly seen in the liver. We present this case to increase awareness of extrarenal angiomyolipomas to be included in the differential diagnosis of a soft tissue mass in various sites and not only in the kidney. Proper and early diagnosis of angiomyolipoma is critical. Angiomyolipomas often present asymptomatic and are found incidental, but with increasing size leads to increasing risk of hemorrhage leading to shock and death.

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