

# Inflammatory Myofibroblastic Tumour of the Colon in a Child: A Case Report and Literature Review

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## Abstract

In the World Health Organization (WHO) classification, inflammatory myofibroblastic tumor is categorized among intermediate neoplasms and can affect various organs such as the lungs, pancreas, bladder, and uterus, while its colonic localization is rare. We present the case of a 4-year-old girl with a history of chronic constipation. Clinical examination revealed the presence of a firm and mobile mass measuring 5 cm in the right hypochondrium. Abdominal ultrasound and thoraco-abdominal CT scan identified a subhepatic mass, accompanied by lymphadenopathy, initially suggestive of lymphoma. Histopathological examination and immunohistochemical studies ultimately established the diagnosis of inflammatory myofibroblastic tumor. Based on this case, we discuss the differential diagnosis and specific histopathological features associated with this rare colonic localization.

## Keywords

Inflammatory Myofibroblastic Tumor, Colon, Inflammatory Pseudotumor, Child, Rare Tumor

## 1. Introduction

Inflammatory Myofibroblastic Tumor (IMT), formerly known as inflammatory

pseudotumor, remains poorly understood and is defined as a distinctive neoplasm. It consists of spindle-shaped myofibroblastic and fibroblastic cells, accompanied by an inflammatory infiltrate comprising plasma cells, lymphocytes, and/or eosinophils [1] [2].

The first observation of IMT dates back to 1905 with Birch-Hirschfeld, and the term inflammatory pseudotumor was introduced by Umiker in 1954 due to its inflammatory and nodular appearance, both clinically and radiologically [3]. This tumor can occur at any age and affects various organs, with a marked preference for the upper and lower respiratory tracts [3].

Localization in the gastrointestinal tract is unusual and is more prevalent in females. Characteristic symptoms in this region include dyspepsia, hypogastric pain, or the discovery of an abdominal mass [4]. Currently, this lesion is classified among tumors of intermediate malignancy; it exhibits a potential for local recurrence (10% to 20%) and a low risk of distant metastases [5]. Diagnosis remains challenging, and surgery is often necessary for complete excision to prevent recurrences that impact the prognosis of this tumor.

Here, we present a new case of IMT adherent to the right colonic angle in a 4-year-old girl, histopathologically confirmed after surgical resection. The report explores the epidemiological, pathological, molecular, and prognostic characteristics of this tumor, along with the challenges associated with its diagnosis.

## 2. Case Presentation

The patient is a 4-year-old girl with a history of chronic constipation since the age of 2, without any other associated signs. In this context, the child's general condition was preserved.

Clinical examination revealed the presence of a firm and mobile mass measuring 5 cm in the right hypochondrium, with no other somatic peculiarities.

Abdominal ultrasound and thoraco-abdominal CT scan highlighted a subhepatic intra-peritoneal mass, accompanied by lymphadenopathy, suggesting a potentially lymphomatous origin (see **Figures 1-3**). Laboratory tests revealed a decrease in hemoglobin levels to 7.2 g/dl and marked thrombocytopenia with platelets at 9200/mm<sup>3</sup>.

A mini laparotomy was performed, revealing a solid, vascularized intraperitoneal mass adherent to the right colonic angle without a cleavage plane. Resection of the colonic portion carrying the tumor mass was carried out.

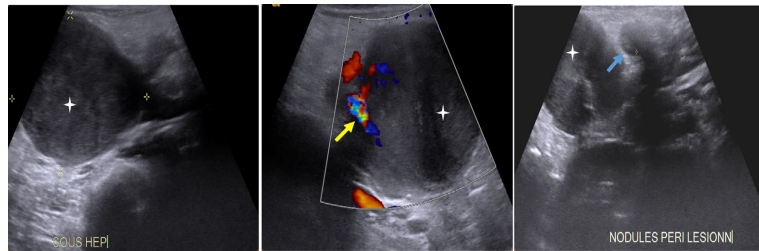
The surgical specimen, a colonic resection of 10 × 2.5 cm adjacent to an exoluminal mass of 5 × 4 × 2 cm, showed on sectioning the presence of a whitish, homogeneous, and well-defined nodular lesion (see **Figure 4**).

Histological analysis confirmed a well-limited myofibroblastic tumor proliferation, composed of long interwoven bundles. The tumor cells were spindle-shaped, with a regular nucleus, fine chromatin, and ill-defined eosinophilic cytoplasm. Mitoses were rare (see **Figures 5-7**).

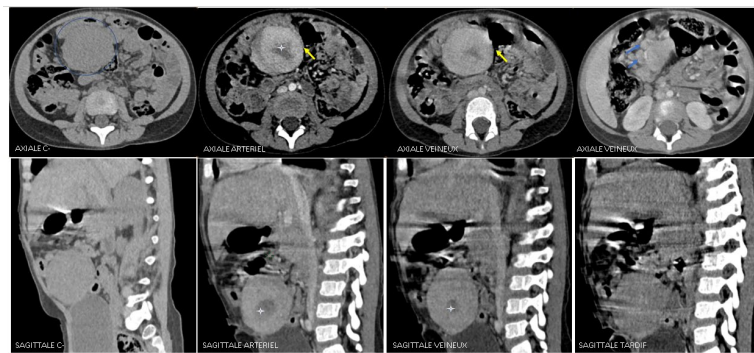
Immunohistochemical study revealed that tumor cells expressed smooth muscle

actin (SMA) and ALK1 while being negative for desmin, myogenin, H-caldesmon, beta-catenin, protein S100, CD117 and Dog1. CD138 marked the plasma cells (see **Figure 8**, **Figure 9**). The clinical, radiological, surgical, and pathological characteristics of the patient are documented in **Table 1**.

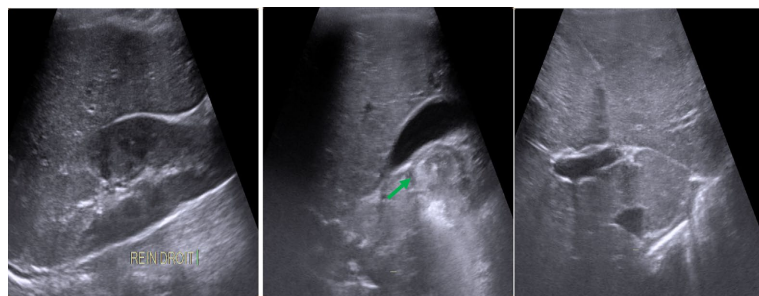
The histopathological study of the surgical specimen confirmed the diagnosis of an inflammatory myofibroblastic tumor. The patient's condition showed a favorable evolution with a two-month follow-up.



**Figure 1. Initial abdominal ultrasound:** shows a subhepatic tissue mass, hypoechoic and heterogeneous (asterisk), rounded, well circumscribed, demonstrating color Doppler flow (yellow arrow), associated with perilesional tissue nodules (blue arrow).



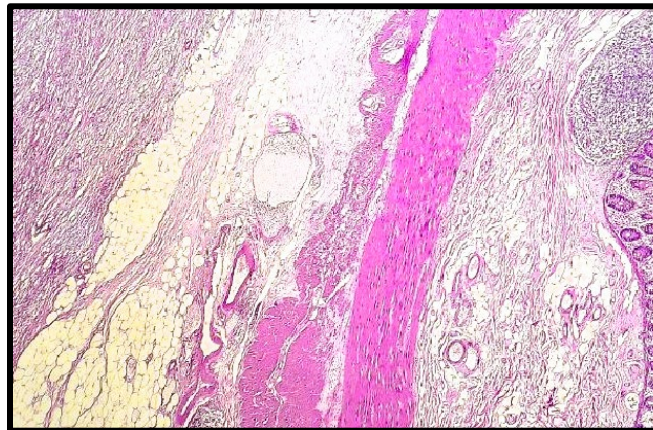
**Figure 2. Abdominal-pelvic CT scan: axial and sagittal sections, in spontaneous contrast and after injection of contrast medium at arterial and venous phases:** Shows the presence of a tissue mass, intraperitoneal, seemingly centered on the mesenteric region, adjacent to the transverse colon (yellow arrows). It is rounded in shape, well circumscribed, isodense in spontaneous contrast (circle), and shows heterogeneous enhancement after contrast (asterisk). There are also some mesenteric lymph nodes present elsewhere (blue arrows).



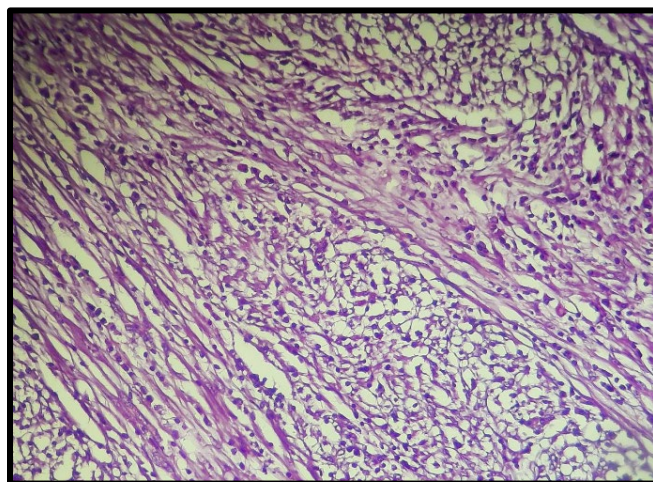
**Figure 3. Follow-up abdominal ultrasound:** no mass syndrome or suspicious lesion in the subhepatic region (green arrow).



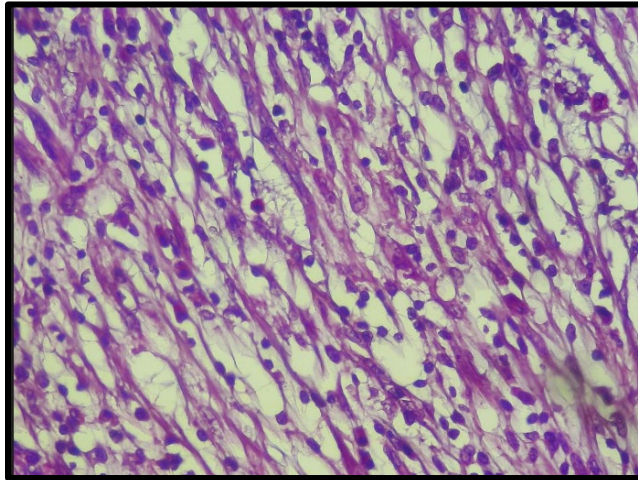
**Figure 4.** Macroscopic examination reveals a well-defined, homogeneous, whitish nodular lesion adjacent to a colonic wall.



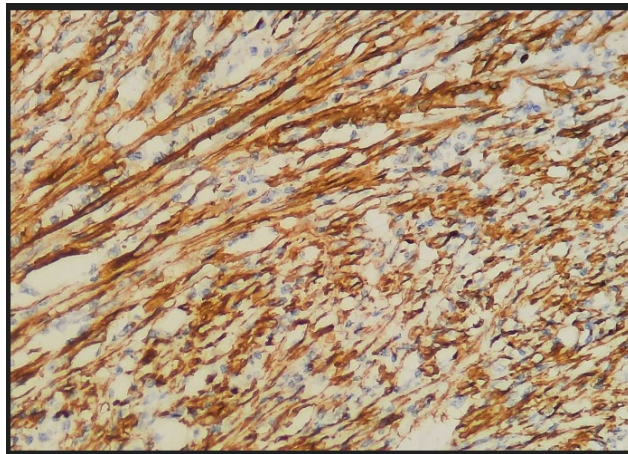
**Figure 5.** Colonic mucosa with storiform myofibroblastic proliferation in the serosa (haematoxylin and eosin  $\times 10$ ).



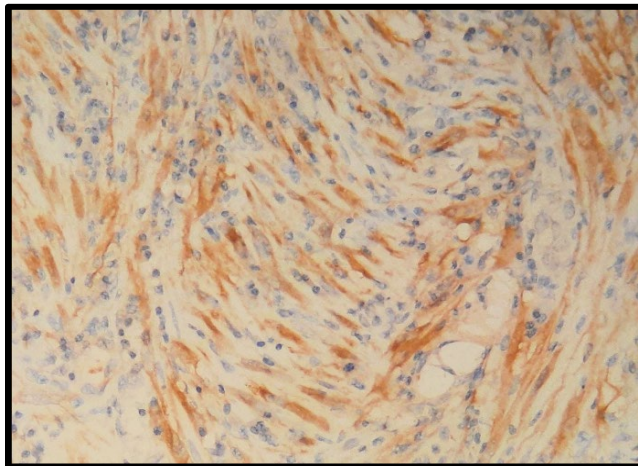
**Figure 6.** Storiform myofibroblastic proliferation associated with a lymphoplasmacytic infiltrate (Hematoxylin and eosin  $\times 20$ ).



**Figure 7.** Storiform myofibroblastic proliferation associated with a lymphoplasmacytic infiltrate (Hematoxylin and eosin  $\times 40$ ).



**Figure 8.** Intense and diffuse cytoplasmic immunostaining with anti-AML antibody (IHC  $\times 400$ ).



**Figure 9.** Intense and diffuse cytoplasmic immunostaining with anti-ALK1 antibody (IHC  $\times 400$ ).

**Table 1.** Clinical, radiological, surgical, and pathological features of the patient.

Parameter	Details
Age/Sex	4-year-old girl
Medical History	Chronic constipation since age 2
General Condition	Preserved, no other associated signs
Clinical Examination	Right hypochondrial mass, firm, mobile, 5 cm
Imaging	Ultrasound and thoraco-abdominal CT: subhepatic mass with lymphadenopathy, suspicious for lymphoma
Laboratory Findings	Hemoglobin: 7.2 g/dL; Platelets: 9200/mm <sup>3</sup>
Surgery	Mini-laparotomy; resection of colonic segment with tumor
Gross Pathology	Colonic specimen 10 × 2.5 cm; exoluminal mass 5 × 4 × 2 cm; whitish, well-defined, homogeneous nodule
Histopathology	Well-circumscribed myofibroblastic proliferation; spindle-shaped cells; rare mitoses
Immunohistochemistry—Positive	SMA, ALK1
Immunohistochemistry—Negative	Desmin, myogenin, H-caldesmon, $\beta$ -catenin, S100, CD117, DOG1
Other Markers	CD138 positive in plasma cells

### 3. Discussion

Inflammatory Myofibroblastic Tumor (IMT) is classified by the World Health Organization (WHO) among intermediate neoplasms and can develop in various organs [6]. Initially described in the lungs, other abdominal-pelvic or head and neck localizations have been reported [7]. Lesions can be located either along the gastrointestinal tract, in the solid organs of the abdominal cavity, or in the retroperitoneum, mesentery, or diaphragm. In the colon, the most common site is the right colon [8]. The prevalence in the colon is relatively low, with only a few cases reported in the literature [9].

IMT mainly affects young adults and children [10]. Its etiology remains unknown, although it has been attributed to several factors including surgical interventions, trauma, autoimmune reactions, as well as infections, particularly those caused by the Epstein-Barr virus and human herpesvirus [11] [12]. Over the past three decades, a subset of these lesions has been found to be neoplastic, with a clonal chromosomal abnormality documented, mainly involving the anaplastic lymphoma kinase (ALK) gene on chromosome 2p23 [10] [11].

The clinical presentation of colonic IMT depends on its anatomical location [11] [13]. Abdominal girth and possibly symptoms of obstruction may be observed. Between 15% and 30% of symptomatic patients present with systemic symptoms such as fever, night sweats and weight loss. [14]. Clinical, hematological, or biochemical manifestations are non-specific and may resolve after surgical resection [15]. Abdominal radiological examinations may reveal the presence of a mass, although its characteristics are not specific [8].

Histopathologically, IMT is a solid tumor primarily composed of spindle cells with a chronic inflammatory component including plasma cells, lymphocytes, and

sometimes histiocytes [8].

On an immunohistochemical level, diffuse cytoplasmic reactivity for vimentin is typical for almost all IMTs. Reactivity for smooth muscle actin and specific muscle actin varies from a focal to a diffuse pattern in the cytoplasm of spindle cells, and desmin is identified in many cases [16]. Confirmation of the presence of the ALK protein (positive in approximately 40% - 60% of cases) is useful to distinguish colonic IMT from other spindle cell neoplasms such as [16]:

- Gastrointestinal stromal tumor (GIST): This tumor may present with lymphocyte-rich inflammation, occasionally with eosinophils or plasma cells. It is positive for DOG1 and CD117, but negative for ALK1 by immunohistochemistry (IHC).
- Inflammatory leiomyosarcoma: Characterized by cigar-shaped nuclei and a more organized fascicular growth pattern.
- Desmoid-type fibromatosis: Identified by spindle cells interwoven with collagen and minimal to no inflammation. It tests negative for ALK1, with a mutation in CTNNB1 (beta-catenin).

Overexpression of interleukin 6 and cyclin D1 has also been described in IMT [17].

IMTs are genetically heterogeneous. In 50% - 60% of cases of IMT in children and young adults, the tumors harbor clonal cytogenetic rearrangements, involving chromosome band 2p23, that fuse the 3' kinase region of the ALK gene with various partner genes [2] [10] [11]. These partner genes include TPM3, TPM4, CLTC, CARS, ATIC, SEC31L1, PPFIBP1, DCTN1, EML4, PRKAR1A, LMNA, TFG, FN1, HNRNPA1, and others [2].

The treatment of choice is based on surgical resection (re-excision of recurrences) and treatment with specific tyrosine kinase inhibitors, such as crizotinib [18].

Generally, the prognosis is favorable, but recurrence may occur in up to 35% of cases, with rare instances of distant metastasis reported (in around 5%) [10] [11] [16]. Adverse factors include an intra-abdominal location and the presence of the epithelioid variant. While there is no established correlation between tumor size, mitotic rate, cellularity, necrosis, and atypia with outcome in conventional IMT, ALK-positive IMTs are less likely to recur and may exhibit less aggressive behavior [16].

#### 4. Conclusions

Colonic Inflammatory Myofibroblastic Tumor (IMT) represents a rare entity, posing significant challenges both diagnostically and clinically. Its key characteristics include myofibroblastic proliferation and variable inflammation, manifesting across a spectrum of macroscopic and microscopic features.

Complete surgical excision remains the cornerstone of colonic IMT treatment. The overall prognosis is generally favorable, with only a few reported cases of malignant transformation, recurrence, or distant metastases. The precise factors in-

fluencing prognosis remain somewhat unclear.

Long-term clinical and radiological follow-up is strongly recommended to enable early detection of any potential recurrence or metastasis. This surveillance approach contributes to ensuring a proactive and tailored management for individual needs of patients with colonic IMT, thereby reinforcing the favorable outlook associated with this rare condition.

## Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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