Cureus
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Open Access Case Report

A Case of Solitary Cecal Neurofibroma: A Rare Incidental Finding

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Review began 09/08/2026
Review ended 10/17/2026
Published 10/23/2026

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DOI: 10.7759/cureus.118426

Abstract

Neurofibromas are benign peripheral nerve sheath tumors that may occur sporadically or in association with neurofibromatosis type 1 (NF1). Gastrointestinal involvement is uncommon, and isolated colonic neurofibroma is particularly a rare entity. We present the case of a 70-year-old man who underwent routine screening colonoscopy and was found to have an incidental 6 mm sessile polyp in the cecum. Histopathological examination demonstrated a spindle cell proliferation involving the submucosa and mucosa. Immunohistochemical staining showed positivity for S-100 and SOX10, while staining for CD117, DOG1, and desmin was negative, supporting the diagnosis of colonic neurofibroma. The patient had no clinical features or family history suggestive of NF1. He remained clinically well during one year of follow-up. Isolated colonic neurofibroma is an exceptionally rare finding that may be discovered incidentally in asymptomatic patients. Recognition of this entity is important because of its potential association with NF1 and the need for appropriate clinical evaluation. This case highlights the rarity of isolated cecal neurofibroma and the importance of histopathological evaluation of incidental colonic lesions identified during routine screening colonoscopy.

Categories: Neurology, Internal Medicine, Gastroenterology
Keywords: cecal polyp, colonic neurofibroma, immuno-histochemical, inflammatory fibroid polyp, intestinal submucosal tumor, s100-positive, screening colonoscopy, solitary neurofibroma

Introduction

Neurofibromas represent benign neoplasms of the peripheral nerve sheath that arise either sporadically or as a manifestation of systemic neurofibromatosis type 1 (NF1) [1]. Neurofibromatosis is a group of genetic disorders that cause noncancerous tumors to grow on nerve tissues, affecting the skin, brain, spinal cord, and sometimes gastrointestinal (GI) organs. Although mesenchymal tumors can occasionally involve the GI tract, an isolated neurofibroma localized strictly to the colon remains an exceedingly rare clinical entity [2,3]. Unlike syndromic, NF1-associated presentations, clinically isolated neurofibromas manifest as solitary findings in individuals without systemic signs, though definitive confirmation of a true sporadic variant requires genetic analysis to exclude germline mutations.

GI involvement is a recognized manifestation in a subset of patients with NF1, whereas isolated variants are exceptionally rare; while often found incidentally, larger or syndromic lesions can present with abdominal pain, bleeding, or abnormal cross-sectional imaging. According to the revised 2021 international consensus criteria, an NF1 diagnosis is established if two or more features are met, such as six or more café-au-lait macules, two or more neurofibromas or one plexiform neurofibroma, axillary/inguinal freckling, optic pathway glioma, two or more Lisch nodules or two or more choroidal abnormalities, characteristic osseous lesions (e.g., sphenoid dysplasia or anterolateral bowing of the tibia), or a heterozygous pathogenic NF1 gene variant. Histopathologically, these lesions characteristically manifest as spindle cell proliferations involving the mucosa and submucosa [4].

Definitive confirmation relies on correlating the histopathological morphology with a targeted immunohistochemical panel, which is crucial for distinguishing neurofibromas from other neural GI polyps like schwannomas [3]. They typically exhibit strong expression of neural crest differentiation markers while remaining entirely negative for GI stromal and myogenic markers to systematically differentiate them from more common mesenchymal mimics. To expand upon the sparse literature surrounding these atypical visceral presentations, we report the case of a sporadic, isolated colonic neurofibroma presenting as an incidental 6 mm sessile cecal polyp discovered during a routine screening colonoscopy in an asymptomatic elderly male patient. The present case is distinctive due to the rare combination of a solitary, diminutive (6 mm), entirely asymptomatic cecal neurofibroma discovered incidentally in a patient with a complete absence of syndromic features.

Case Presentation

A 70-year-old male patient was referred to our gastroenterology clinic for screening colonoscopy. He denied any heartburn, nausea, vomiting, or abdominal pain. He was otherwise healthy and also denied having

blood in his stool, fever, or unintentional weight loss. His past medical history was significant for asthma, chronic obstructive pulmonary disease (COPD), and Barrett's esophagus. His home medications included an albuterol inhaler, pantoprazole, and multivitamins. He had no family history of GI or any other malignancy. He had a 40 pack-year smoking history.

Physical examination revealed obesity (BMI: 36 kg/m^2) and a soft abdomen with no tenderness or palpable mass. A thorough, full-body mucocutaneous clinical examination under optimal lighting revealed an absolute absence of café-au-lait spots, cutaneous neurofibromas, or intertriginous freckling. Cardiovascular, neurological, and dermatological system examination was unremarkable (Table *1*).

Organ System	Physical Examination Findings
Cardiovascular	Regular rate and rhythm - Normal S1 and S2 - No murmurs, rubs, or gallops appreciated - Peripheral pulses intact.
Neurological	Alert and oriented to time, place, and person - No focal neurological deficits.
Dermatological	No café-au-lait macules - No cutaneous neurofibromas - No axillary or inguinal freckling - No other cutaneous lesions identified.

TABLE 1: Physical examination findings

The respiratory examination was consistent with underlying asthma and COPD. Routine laboratory results were largely unremarkable, including hematologic, renal, hepatic, and coagulation parameters. Hepatitis serology was negative (Table *2*).

Laboratory Test	Patient Value	Reference Range
Hemoglobin	13.9 g/dL	13.5–17.5 g/dL
MCV	101 fL	80–100 fL
WBC count	8,100 /µL	4,000–11,000 /µL
Neutrophils	6.4 ×10³ /µL	2.0–7.5 ×10³ /µL
Platelets	251,000 /µL	150,000–450,000 /µL
Creatinine	0.9 mg/dL	0.6–1.3 mg/dL
Glucose (random)	116 mg/dL	70–140 mg/dL (random)
ALT	22 U/L	9–46 U/L
AST	24 U/L	10–40 U/L
ALP	71 U/L	35–130 U/L
Prothrombin Time (PT)	12.2 seconds	11.3–13.5 seconds
aPTT	29.7 seconds	25.0–35.0 seconds
INR	1.0	0.8–1.1
HBsAg	Negative	—
anti-HCV	Negative	—

TABLE 2: Laboratory findings

MCV = mean corpuscular volume; WBC = white blood cell; ALT = alanine aminotransferase; AST = aspartate aminotransferase; ALP = alkaline phosphatase; aPTT = activated partial thromboplastin time; INR = international normalized ratio; HBsAg = hepatitis B surface antigen; HCV = hepatitis C virus

As part of routine screening, colonoscopy was performed up to the cecum. A 6 mm sessile polyp was

identified in the cecum (neurofibroma) (Figure 1). On endoscopic evaluation, the lesion appeared as a smooth, sessile mucosal nodule with a normal, regular overlying vascular and pit pattern; tactile interrogation with biopsy forceps demonstrated a firm consistency. The lesion was subsequently removed entirely via cold snare polypectomy without complications, with histopathological review confirming complete excision and negative resection margins. Other findings were left-sided diverticulosis of the colon and grade 1 internal hemorrhoids.

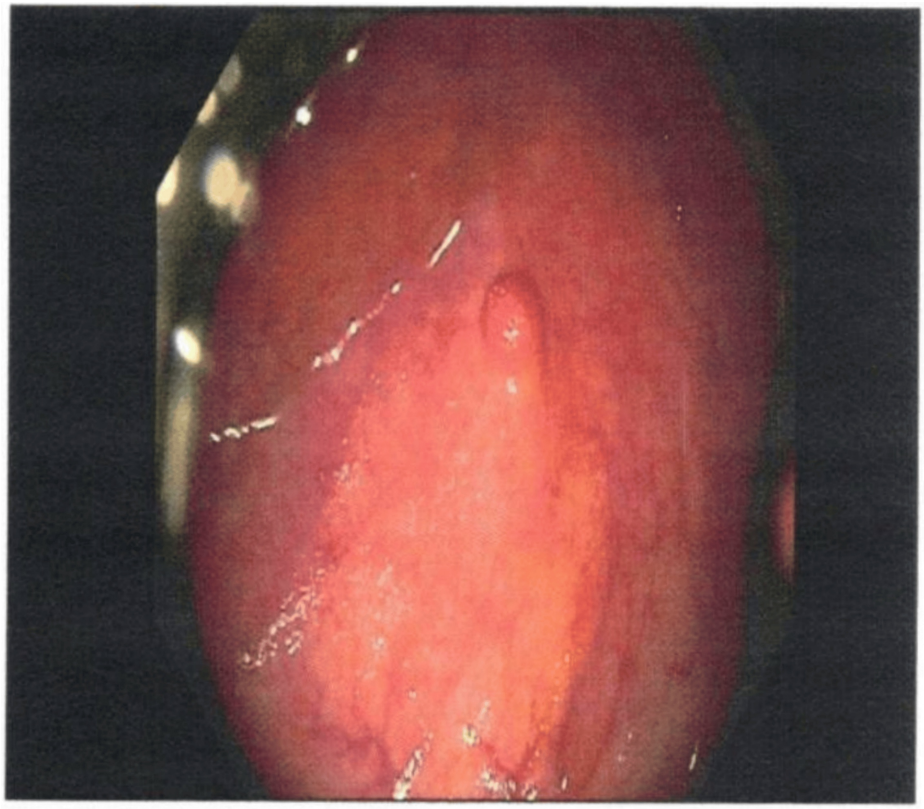


FIGURE 1: Endoscopic view of sessile polyp in cecum

Histologically, the cecal polyp revealed spindle cell proliferation in the submucosa extending into the mucosa (Figure 2). This histopathological evaluation (H&E, ×100) of the tissue sections revealed a benign, well-circumscribed mucosal lesion expanding within the lamina propria, composed of a loose proliferation of uniform, spindle-shaped cells with characteristic wavy nuclei that gently displaced the adjacent normal colonic crypts without causing architectural destruction, cellular atypia, or mitotic activity.

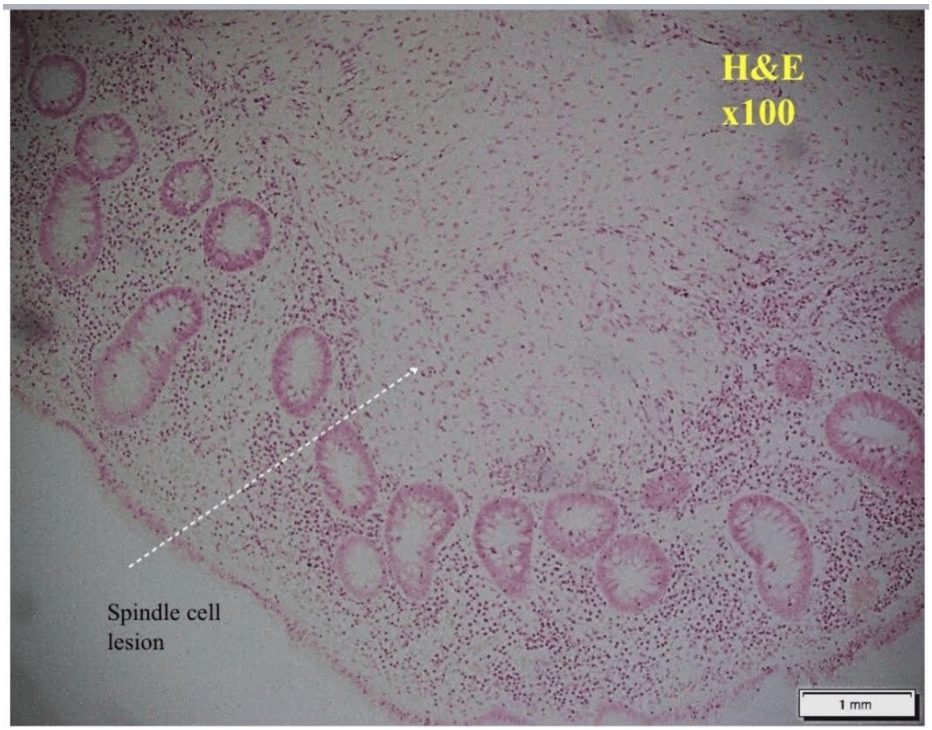


FIGURE 2: Histopathology of cecal neurofibroma.

H&E stain at low-power magnification (×100) showing a well circumscribed lesion composed of uniform spindle cells with characteristic wavy nuclei involving the mucosa and submucosa, gently displacing adjacent normal colonic crypts.

Subsequently, immunohistochemistry showed tumor cells were reactive to S-100, SOX10, and CD68 and nonreactive to CD117, DOG1, and desmin. These findings were consistent with a colonic submucosal neurofibroma, a rare entity in the colon in the absence of systemic neurofibromatosis. The patient continued to receive outpatient follow-up and remained free of clinical features suggestive of neurofibromatosis at the one-year follow-up.

Discussion

Our patient was diagnosed with an isolated colonic neurofibroma, which is an exceptionally rare entity. Intestinal involvement is a recognized manifestation in a subset of patients with NF1, though its documented incidence varies substantially depending on the population evaluated. Most lesions are found in the upper GI tract and less frequently in the lower GI tract [2].

Neurofibromas may be solitary, multiple, or plexiform. They are mostly multiple at presentation and are part of a hereditary disorder with two clinical forms: NF1 and NF2 [3]. To date, solitary neurofibromas localized strictly to the large bowel remain exceptionally rare, with only a limited number of cases documented over the past two decades (Table *3*). A comprehensive literature search was conducted across the PubMed database for English-language articles published between 2000 and 2026. The search utilized combinations of medical subject headings (MeSH) and keywords, including 'isolated colonic neurofibroma,' 'sporadic colonic neurofibroma,' and 'cecal neurofibroma.' To ensure completeness, case selection criteria were strictly restricted to peer-reviewed, pathologically confirmed solitary neurofibromas confined entirely to the large bowel, while syndromic (NF1-associated) or multiple GI nerve sheath tumors were excluded.

Case Report (Author, Year)	Age	Sex	Presentation	Location of neurofibroma	Pathology
Bononi et al. (2000) [5]	68	F	Rectal bleeding, tenesmus	Diffuse colon involvement	Multiple tumors; S100(+), SMA(-)
Panteris et al. (2005) [6]	65	F	Bloody diarrhea, abdominal pain	Descending colon	Neurofibroma; S100(+), Vimentin(+), etc.
Carter and Laurini (2008) [7]	52	F	Non-bloody diarrhea, pain	Rectum & transverse colon	Neurofibroma; S100(+)
Hindy et al. (2012) [8]	59	M	Asymptomatic	Transverse colon	Neurofibroma; S100(+); 3 mm
Chelimilla et al. (2013) [3]	70	F	Asymptomatic	Ascending colon	Neurofibroma; S100(+), CD117(-), SMA(-), Desmin(-)
Ahn et al. (2016) [9]	26	F	Asymptomatic	Sigmoid colon	Neurofibroma; S100(+), CD34(+), c-Kit(-), DOG1(-)
Fulgence et al. (2016) [10]	37	F	Abdominal mass, abdominal pain	Transverse colon	Neurofibroma; no IHC
Adioui et al. (2018) [11]	29	F	Left iliac fossa pain	Sigmoid colon	Neurofibroma; S100(+), CD34(-), c-Kit(-)
Miao et al. (2018) [12]	24	F	Pain, mass in stool	Ileocecal valve	Neurofibroma; S100(+), low Ki-67
Ogundoyin and Ajani (2018) [13]	12	M	Painless mass	Splenic flexure	Neurofibroma; no IHC
Imagami et al. (2020) [14]	81	F	Asymptomatic	Hepatic flexure	Neurofibroma; S100(+), CD34(-)
Ghoneim et al. (2020) [2]	51	M	Asymptomatic	15 cm from anal verge	Neurofibroma; S100(+), low Ki-67
Sun et al. (2020) [15]	33	F	Asymptomatic	Ascending colon	Neurofibroma; S100(+)
Tinguria (2022) [16]	76	M	Anemia, GERD	Sigmoid colon	Neurofibroma; SOX10(+), S100(+), CD34(+)
He et al. (2024) [17]	55	F	Abdominal pain, bleeding	Distal rectum	Spindled neurofibroma; SOX10(+), S100(+)
Present case (2026)	70	M	Asymptomatic	Cecum	S100(+), SOX10(+), CD68(+), CD117(-), desmin(-)

TABLE 3: Clinical and pathological characteristics of documented cases of isolated colonic neurofibroma (2000-2026)

F = female; M = male; GERD = gastroesophageal reflux disease; IHC= immunohistochemistry

A distinct anatomical predilection for the left colon is observed, particularly the rectum and sigmoid colon [9,11]. Conversely, right-sided lesions, specifically those localized to the cecum and ascending colon, remain exceedingly rare [3,12,15]. While the review of published cases suggests right-sided lesions frequently present as asymptomatic incidentalomas during routine endoscopic screening [3,15], mirroring the present case, this trend must be interpreted with caution due to the small sample size currently available in the literature. However, atypical presentations exist. While isolated colonic neurofibromas can occasionally manifest as symptomatic abdominal masses [10], Miao et al. described a novel case originating near the ileocecal valve that spontaneously auto-amputated and passed per anus [12]. This anatomical distribution highlights the exceptional rarity of cecal localization and underscores the clinical significance of our case as a completely asymptomatic, sporadic cecal neurofibroma identified during routine screening colonoscopy.

Dense plexiform neurofibromas may infiltrate the mesentery and lead to vascular or neural compromise. Clinically, patients may present with rectal bleeding, abdominal pain, palpable abdominal mass, nausea, vomiting, or features of obstruction due to intussusception or extrinsic compression. Unlike the case reported by He et al., where the patient presented with abdominal pain and bleeding due to distal rectal involvement [17], our patient remained completely asymptomatic and was diagnosed incidentally during routine screening colonoscopy. Approximately 50% of patients with neurofibromas have a family history of the disease, while the remainder represent sporadic mutations [1]. In our case, there was no family history of

neurofibromatosis or GI malignancy, which made the diagnosis more challenging.

Macroscopically, solitary colonic neurofibromas typically present as smooth, sessile polyps that mimic standard polyps, though physical interrogation with forceps often reveals a distinct firm consistency that can raise clinical suspicion. While a formal diagnostic protocol for isolated lesions is not universally established, a practical clinical approach involves a thorough full-body mucocutaneous exam and detailed family history to screen for syndromic features. In patients with an entirely negative systemic presentation and family pedigree, advanced genetic sequencing or cross-sectional neuroimaging are typically deferred, with follow-up focusing on standard post-polypectomy surveillance.

The differential diagnosis for neurofibroma of the GI tract includes schwannoma, myxoma, inflammatory fibroid polyp, and GI stromal tumor (GIST). Histopathological examination remains the gold standard for diagnosis. Neurofibromas typically demonstrate diffuse S-100 positivity, in contrast to myxomas, which lack this expression. Schwannomas and inflammatory fibroid polyps can be distinguished based on histologic architecture and immune profile, including absence of calretinin expression and lack of a peripheral lymphoid cuff, respectively [18,6]. Neurofibromas may also show CD34 and factor XIIIa positivity, in contrast to schwannomas. Importantly, neurofibromas are negative for CD117 and DOG1, whereas GISTs typically show strong positivity for these markers [18].

An important clinical teaching point in the evaluation of GI neurofibromas is the utility of a formal ophthalmic slit-lamp examination to screen for Lisch nodules; however, their absence does not definitively rule out underlying NF1, particularly given the updated inclusion of choroidal abnormalities in the revised consensus criteria 2021 [4]. Although a slit-lamp examination was not done in the present case due to a completely negative family history and the absence of any cutaneous or systemic syndromic markers, clinicians should consider this non-invasive screening tool when underlying hereditary syndromes are suspected. Documenting a thorough mucocutaneous and ocular workup remains critical to distinguishing truly sporadic, isolated colonic neurofibromas from syndromic variants like NF1 or multiple endocrine neoplasia type 2B (MEN2B). To confirm the diagnosis of a solitary colonic lesion, systemic manifestations of NF1 must be carefully excluded. Patients with classical NF1 often require neuroimaging to evaluate characteristic central nervous system lesions, including brain and spinal cord involvement [19]. In the present case, the patient had no neurological symptoms or clinical evidence of NF1, supporting a sporadic isolated lesion.

Malignant transformation in NF1 has been reported, particularly in larger lesions and plexiform subtypes. These malignant peripheral nerve sheath tumors are aggressive and associated with poor prognosis, with 5- to 10-year overall survival rates historically ranging from 52% to 34%, respectively [20]. Prognosis depends on tumor size, anatomical location, stage, presence of metastasis, and completeness of surgical resection.

A comprehensive clinical evaluation is essential to exclude NF1 due to its variable and sometimes subtle presentation [21]. In some cases, GI manifestations may represent the initial presentation of the disease. While our clinical workup strongly supports a sporadic diagnosis, certain diagnostic limitations must be acknowledged. First, while histomorphology aligned with a benign neural lesion, our immunohistochemical panel was targeted rather than exhaustive. Second, the classification of this lesion as entirely sporadic relied strictly on a comprehensive clinical, morphological, and histopathological evaluation, as advanced genetic testing and systemic neuroimaging were not clinically indicated or performed. However, given the rarity of isolated colonic neurofibroma, there are currently no established guidelines regarding long-term surveillance or optimal colonoscopic follow-up strategies, whether using conventional colonoscopy or CT colonography.

Conclusions

Solitary colonic neurofibroma, particularly when involving the cecum, is a rare GI lesion that is most often detected incidentally during routine colonoscopy. Histopathological examination with appropriate immunohistochemical evaluation is essential for confirming the diagnosis and distinguishing it from other colonic lesions. Although GI neurofibromas may be associated with NF1, a clinically isolated colonic neurofibroma can present without systemic manifestations, as demonstrated in this case, though definitive molecular exclusion of underlying disease is limited without germline genetic assessment.

Given the low incidence of this condition, evidence regarding long-term surveillance and follow-up remains limited, and there are currently no well-established guidelines for the optimal management or colonoscopic follow-up of patients with isolated cecal neurofibroma. Further studies and accumulation of reported cases are needed to better define appropriate long-term surveillance strategies for this rare entity. This case contributes to the limited literature on isolated cecal neurofibroma and underscores the importance of including this entity in the differential diagnosis of incidental colonic polyps.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Shuja Ur Rehman, Jack Husney

Acquisition, analysis, or interpretation of data: Shuja Ur Rehman, Jack Husney

Drafting of the manuscript: Shuja Ur Rehman, Jack Husney

Critical review of the manuscript for important intellectual content: Shuja Ur Rehman, Jack Husney

Supervision: Shuja Ur Rehman, Jack Husney

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **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 have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

Acknowledgements

Additional clinical details including endoscopic images and immunohistochemical findings are available from the corresponding author upon request at shujarehmanus@gmail.com

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