

The Isolated Rectal Neurofibroma: A Rare Case Report

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Received date: October 04, 2016; Accepted date: November 07, 2016; Published date: November 11, 2016

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Abstract

Gastrointestinal involvement is noted in up to 25% of patients with neurofibromatosis. Most neurofibromas of the gastrointestinal tract are noted in the stomach and duodenum, the colon can be involved rarely. Most of these occur as a part of colonic involvement in NF1. Solitary neurofibroma of the colon is extremely rare. We present the case of a 13-year-old female having an isolated rectal neurofibroma without any systemic signs and family history of neurofibromatosis; very few numbers of such cases have been reported. The clinical significance of detecting these cases lies in the need of follow-up of these patients as it can be associated with or it can be the initial presentation of neurofibromatosis and there lies a risk of malignant transformation.

Keywords: Neurofibromatosis; Intestinal; Rectal lesion; Neurofibroma

Introduction

Solitary gastrointestinal neurofibroma is a very rare presentation. The clinical manifestations of gastrointestinal neurofibroma vary, being completely asymptomatic to massive lower gastrointestinal bleeding requiring aggressive management. The final diagnosis completely relies on histopathological examination following either surgical resection of the involved bowel or colonoscopic removal of the polyps.

Case Report

A 13-year-old female presented with rectal bleeding. Her medical history was not significant. The patient did not have any significant family history of neurofibromatosis or gastrointestinal malignancies. On physical examination the patient had an average built. There was no tenderness or palpable masses present on abdominal examination. Head, neck, lung, skin, abdominal and neurological examinations were unremarkable. Laboratory data showed a hemoglobin level of 9.3 g/dL, a white blood cell count of 10600/ μ L, a platelet count of 8,19,000/ μ L and a serum creatinine of 0.9 mg/dL. Basic metabolic profile, liver function tests, and coagulation profile within normal limits. Hepatitis and HIV serology was negative. MRI pelvis was performed which showed lobulated mass arising from posterior wall of rectum. On colonoscopy up to the caecum, there was a large noduloproliferative mass from 2 cm to 10 cm from anal verge was noted. Biopsy was taken. Microscopically, section shows spindle cell proliferation. Immunohistochemical stains were performed and were suggestive of neurofibroma.

The patient was operated, when a low anterior resection was done. The patient continued to be followed up in our outpatient clinic and has not developed any systemic signs of neurofibromatosis even after 15 months of follow-up.

MRI pelvis was suggestive of 72 × 58 × 73 mm sized lobulated altered signal intensity lesion arising from the posterior wall of rectum.

The lesion appeared iso intense on T1w, heterogeneously hyper intense on T2w, does not suppress on STIR and showing intense post contrast enhancement. Urinary bladder was compressed anteriorly due to mass effect (Figures 1a–1f).

Histopathologically, the noduloproliferative lesion showed bland spindle cell proliferation in the submucosa. Immunohistochemical stains stated that the tumor cells were immunoreactive to S100 and vimentin protein (Figures 2a–2c) and were non-reactive to CD117 and desmin antibodies. These findings were consistent with a diagnosis of rectal submucosal neurofibroma.

Discussion

Neurofibromas are benign neoplasms characterized by the proliferation of neural and connective tissue components of peripheral nerves like Schwann and perineural cells and myofibroblasts. S100 protein immunostaining confirms neurofibromas [1]. The differential diagnosis of neural lesions of the colon comprises of neurofibromas, ganglioneuromas, schwannomas, perineuriomas and granular cell tumors [2]. Neurofibromas can be solitary, multiple or plexiform. They are usually multiple upon presentation and are part of an autosomal dominant hereditary disorder with two clinical entities: neurofibromatosis type 1 (von Recklinghausen neurofibromatosis or peripheral neurofibromatosis; NF1) and neurofibromatosis type 2 (central neurofibromatosis or bilateral acoustic neurofibromatosis; NF2). These disorders have a wide range of clinical manifestations involving the skin, nervous system, eyes, bones, gastrointestinal tract other body parts. Gastrointestinal involvement is uncommon with only 25% of patients with NF1 reported to have gastrointestinal involvement, mostly involving the stomach or small bowel [3].

There are two pathological types of gastrointestinal involvement present; one is ganglioneuromatosis in which hyperplasia and hypertrophy of the nerve plexuses and ganglionic cells in the mucosa, other is neurofibromatosis in which hyperplasia of neuronal cells in the submucosa, muscularis propria or even from the serosa. Gastrointestinal stromal tumors with different degrees of neuronal and smooth muscle differentiation and carcinoid tumors of the perianipullary region can be present as associated findings [4–6].

Intestinal neurofibromas generally involve sub mucosa but may extend to the serosa. Diffuse involvement in the form of plexiform neurofibromatosis of the mesentery or retroperitoneal space may cause arterial compression or nerve injury. Although clinical manifestations are non-specific with gastrointestinal neurofibromatosis, some patients may have abdominal pain, constipation, anemia, blood in stool and an abdominal mass. Intestinal or biliary obstruction, ischemic bowel, perforation, intussusception, and mega colon are some serious complications reported. Neurofibromas of the colon are extremely rare and usually, occur in association with NF1. Isolated neurofibromas without any other systemic signs of neurofibromatosis are even rare. There are approximately 10 cases of isolated colon neurofibromas were reported. Most of these cases were female. The age range at diagnosis was 45–70 years. There is a slight predilection for the rectal involvement. Our patient is an example of the same but presents at the

age of 13 which is very rare [7]. Sometimes, solitary gastrointestinal neurofibromas may be the initial sign of NF1 in patients without any other clinical manifestations of the disease.

Solitary neurofibromas have also been reported in mesentery [8], the ileum [9], retroperitoneum, the anal canal [10], the oesophagus [11], the soft palate [12], the gallbladder [13] and the common bile duct [14]. These lesions are typically benign, but have an increased risk of malignant transformation, particularly when association with NF1 and in larger, plexiform lesions [15]. Very few cases of isolated colon neurofibroma have been reported. However, close clinical follow-up of these patients is needed as isolated bowel neurofibroma could be the initial sign of neurofibromatosis and its associated risk of malignant transformation.

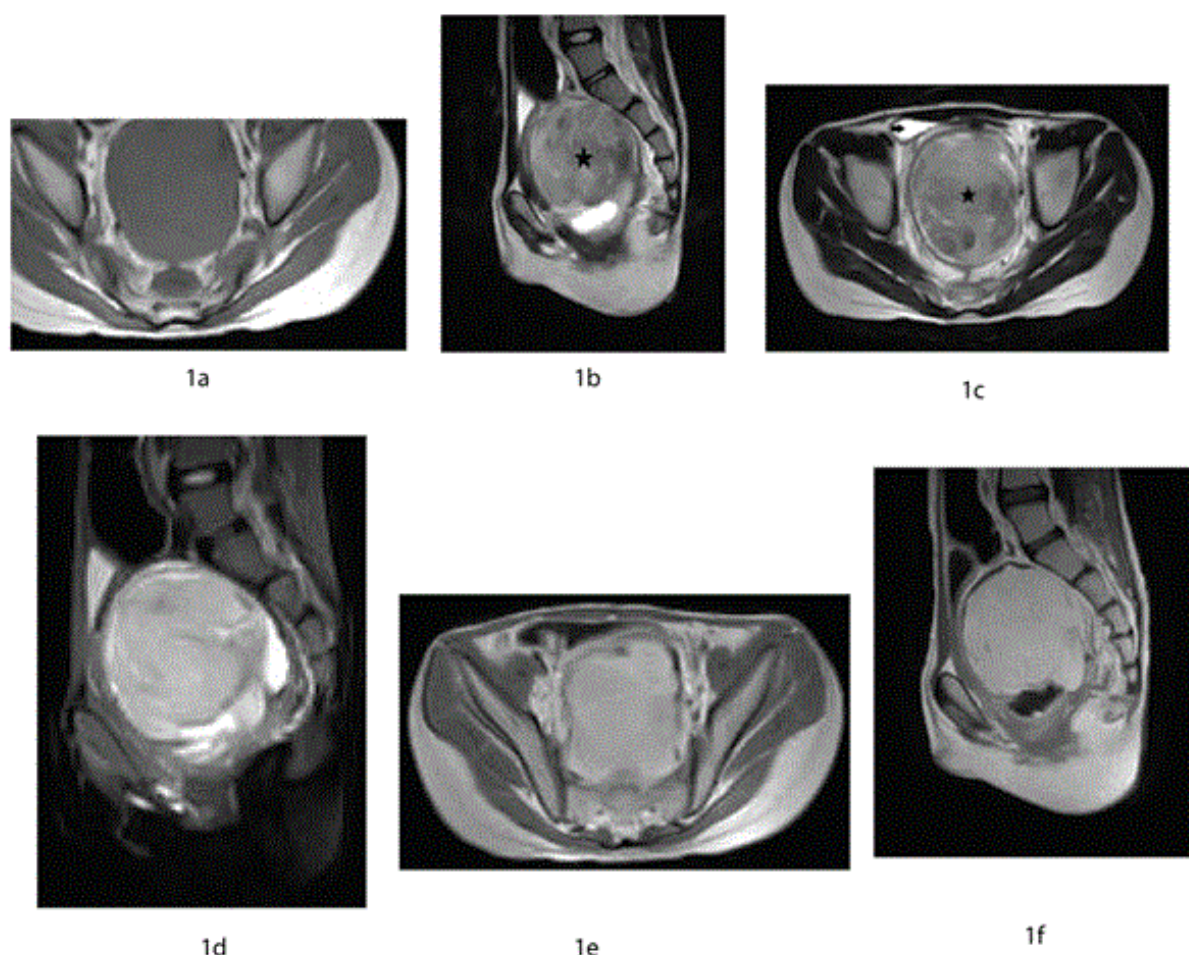


Figure 1: axial T1W(1a), sagittal T2w(1b), axial T2w(1c), STIR sagittal(1d), T1w post contrast axial(1e) and sagittal(1f) images show lobulated isointense on T1W, heterogeneously hyper intense on T2w and bright on STIR and intensely enhancing on post contrast images lesion (*) arising from the posterior wall of the rectum compressing urinary bladder anteriorly (→).

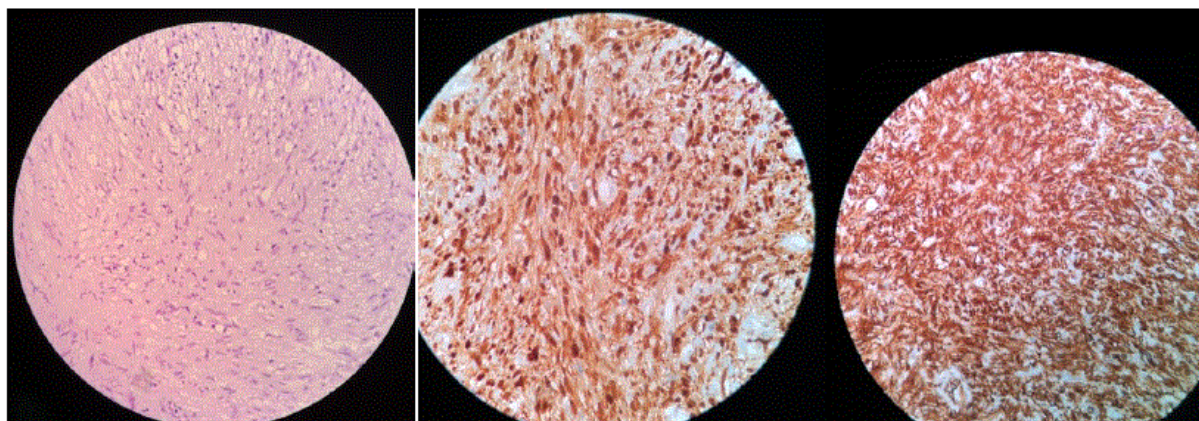


Figure 2: (2a)Rectal lesion showing bland cell proliferation in sub mucosa (2b and 2c) immunohistochemical stains show the tumor cells are strongly positive for S100(2b) and Vimentin protein (2c).

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