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Surgical management of an appendiceal neurofibroma in a neurofibromatosis-1 patient: A case report

Averi Wilson [a], John Slopis [b], Richard Andrassy [c], Sadhna Dhingra [d], Mary Austin [a,e,f,*]

[a] Department of Surgical Oncology, The University of Texas MD Anderson Cancer Center, Houston, TX, USA
[b] Department of Neuro-Oncology, The University of Texas MD Anderson Cancer Center, Houston, TX, USA
[c] Department of Surgery, The University of Texas Medical School at Houston, Houston, TX, USA
[d] Department of Pathology, The University of Texas MD Anderson Cancer Center, Houston, TX, USA
[e] Department of Pediatrics, Children's Cancer Hospital, The University of Texas MD Anderson Cancer Center, Houston, TX, USA
[f] Department of Pediatric Surgery, The University of Texas Medical School at Houston, Houston, TX, USA

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ABSTRACT

Neurofibromatosis-1 (NF1) is an autosomal dominant disease with an incidence of 1 in every 2500 to 3000 births [1]. Although neurofibromas that result from the NF1 gene mutation are typically benign, they may cause complications by compressing neighboring structures or undergoing malignant transformation. The various manifestations and symptoms of NF1 may cause a number of complications throughout childhood and into adulthood, and patients often require surgical management of these complications. A young adult with a previous NF1 diagnosis presented with symptoms concerning for appendicitis. Subsequent imaging and biopsy revealed a rare appendiceal neurofibroma. The patient underwent an ileocecectomy and pathology confirmed the diagnosis of appendiceal neurofibroma. The variety of symptoms and complications associated with NF1, such as vascular abnormalities and airway obstruction, create unique surgical risks for these patients. These risks should be understood and accounted for by surgeons who may encounter NF1 patients.

Neurofibromatosis-1, an autosomal dominant disease with an incidence of 1 in 2500 to 1 in 3000 births, is typically characterized by multiple benign cutaneous, subcutaneous, or plexiform neurofibromas [1]. NF1 patients are typically diagnosed during childhood, require multiple operations both as children and as adults, and are followed by a child neurologist throughout their lives. Complications associated with NF1 are often a result of the spatial arrangement of neurofibromas or result from other consequences of the gene mutation, such as scoliosis; however, malignant transformation of neurofibromas may occur in some patients. Close monitoring, clear imaging, and precise operative management are important in the care of NF1 patients in order to reduce complications associated with both benign and malignant masses. Here, we present a case of an NF1 patient with a rare appendiceal neurofibroma and consider the unique surgical risks associated with NF1.

1. Case report

A twenty-four year old young man presented to an emergency department with complaints of abdominal pain, nausea, and vomiting suggestive of acute appendicitis. The patient's medical history was significant for NF1 and hypertension secondary to left renal artery stenosis. Hypertension was first noted at three years of age and subsequent CT angiogram revealed multiple anomalous renal arteries, one of which was stenotic and required a stent (Fig. 1). These vascular abnormalities, along with the presence of café-au-lait macules and cutaneous fibromas led to the diagnosis of NF1 at seven years of age. He also suffers from developmental delay and aggressive behavior, both of which may be attributed to NF1.

Upon physical examination in the emergency department, multiple cutaneous fibromas and café-au-lait macules (Fig. 2), as well as tenderness to palpation in the right lower quadrant of the abdomen, were noted. The patient was afebrile. Based on CT

* Corresponding author. 1400 Pressler, Unit 1406, Houston, TX 77030-1439, USA. Tel.: +1 713 794 1345.
E-mail address: maustin@mdanderson.org (M. Austin).

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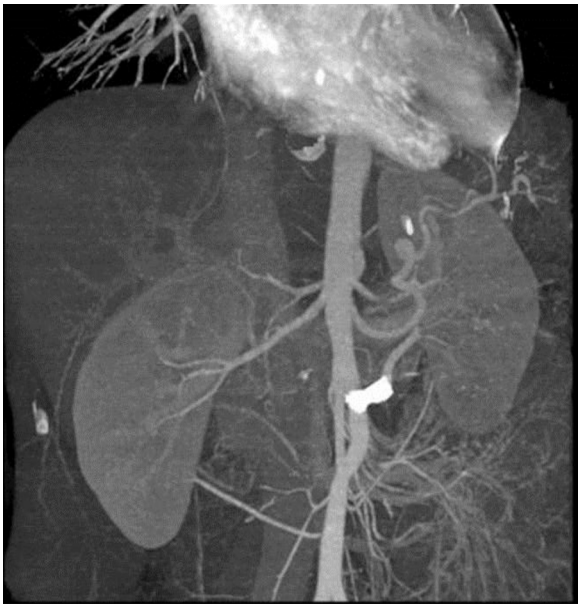

Fig. 1. CT angiogram. Note multiple anomalous renal arteries and a stent placed in a left renal artery.

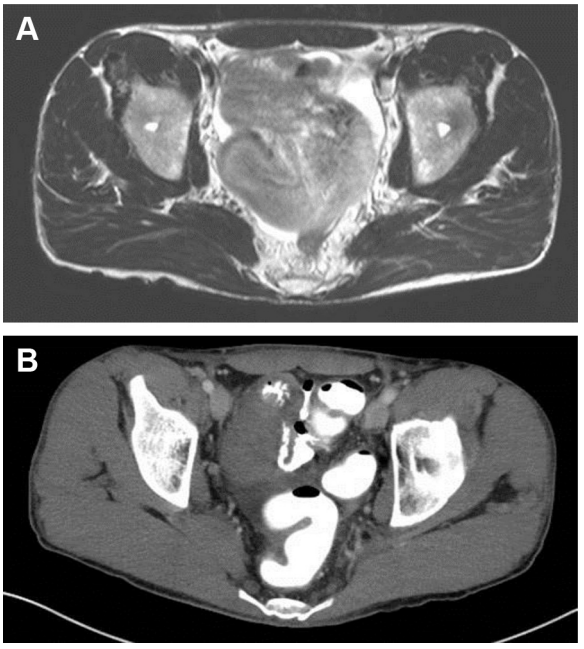

Fig. 3. (A and B) MRI with contrast of the abdomen and pelvis demonstrating a T2 hyperintense elongated appendiceal mass (top) and CT with contrast of the abdomen and pelvis demonstrating a mass-like enlargement of the appendix (bottom).

findings, the patient was diagnosed with appendicitis and possible mesenteric panniculitis and he was started on intravenous cefotetan. He was discharged five days later after symptoms resolved. Further review of CT findings raised concern of an associated appendiceal mass, and follow-up imaging demonstrated a mid-appendiceal mass of 2.5 cm × 7 cm (Fig. 3A and B).

An endoscopic biopsy was performed and revealed a positive S100 immunostain consistent with appendiceal neurofibroma (Fig. 4).

The patient then underwent a hand-assisted laparoscopic ileocecectomy. There were no perioperative complications and the patient was discharged the following day.

Pathological assessment revealed a 8.0 cm circumferential intramural appendiceal mass located within 3.5 cm of the cecal margin (Fig. 5). Histological sectioning demonstrated a plexiform neurofibroma extending from the mucosa to the muscularis propria (Fig. 6A). There was no nuclear atypia or signs of progression to a malignant peripheral nerve sheath tumor. S100 and neurofilament immunostains were positive, confirming the diagnosis of appendiceal neurofibroma (Fig. 6B). CD117 was negative, excluding gastrointestinal stromal tumor (GIST) as a possible diagnosis.

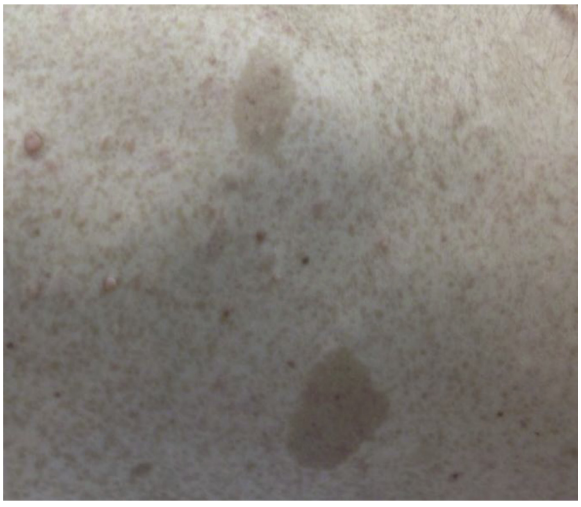

Fig. 2. Café-au-lait macules.

2. Discussion

Neurofibromatosis-1 is estimated to effect 1 in every 2500–3000 births [1]. NF1 occurs as the result of an autosomal dominant mutation in the NF1 tumor suppressor gene [2]. Clinical diagnostic criteria for NF1 have been established (Table 1), but genetic testing is necessary in patients who are not easily diagnosed by clinical criteria or who were previously diagnosed with NF1 and wish to participate in family planning counseling.

Although neurofibromas are typically benign, children and adults with NF1 are at risk for malignant transformation. Neurofibromatosis-1 (NF1) patients have a lifetime risk of malignancy 3% greater than the general population [4]. Specifically, adult and pediatric patients are at risk of glioma and malignant schwannoma, and children with NF1 are at an increased risk for mylogenous leukemia and rhabdomyosarcoma [5]. Tumors such as carcinoid and malignant peripheral nerve sheath tumors also arise at an increased frequency in NF1 patients and often have a poor prognosis [4]. More common benign neurofibromas can also cause a variety of complications. NF1 patients are at risk for optic pathway gliomas, neural tumors, gastrointestinal tumors, vascular abnormalities such as renal stenosis and anomalous vessels, orthopedic issues such as scoliosis, and cosmetically disfiguring lesions [6]. Thus, NF1 patients often require surgical resection of their tumors, including both benign and malignant lesions.