

Case Report

A rare case of a solitary perianal neurofibroma

Dmitriy Kim^{1*}, Elizabeth George², Maheshwari Nalla², Subin Chirayath²

¹Department of General Surgery, Flushing Hospital Medical Center, Flushing, New York, United States

²Department of Surgery, New York Institute of Technology College of Osteopathic Medicine, Old Westbury, New York, United States

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*Correspondence:

Dr. Dmitriy Kim,
E-mail: kdv1982@gmail.com

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ABSTRACT

Neurofibromas are benign nerve sheath tumors commonly associated with Neurofibromatosis type 1 (NF1). In patients without the diagnosis of the NF1, these lesions are referred to as solitary neurofibromas. Gastrointestinal involvement is exceedingly rare in NF1 and can manifest in a variety of different ways. This case report involves a 50-year-old hispanic female with an isolated neurofibroma of the perianal region in the absence of any medical or family history of NF1. Since there are very few cases of solitary neurofibromas reported in literature, it is important to document such cases and follow patients with this disease closely given that it can be an initial presentation of NF1 and because of its possible malignant potential.

Keywords: Neurofibromatosis type 1, Neurofibromas, Genetic disorders, Von Recklinghausen's disease

INTRODUCTION

Neurofibromas are benign nerve sheath tumors commonly associated with Neurofibromatosis type 1 (NF1). This is an autosomal dominant inherited disorder of the NF1 gene on chromosome 17. NF1 is a known tumor suppressor gene and is involved with central nervous system development. This can lead to multiple manifestations, including neurofibromas, cafe au lait spots, and neuropsychological behavioral abnormalities.¹

The diagnostic criteria for NF1 requires 2 out of 7 primary signs including at least 6 cafe au lait spots, at least 2 axillary freckles, at least 2 neurofibromas, optic nerve glioma, at least 2 iris hamartomas, pseudoarthrosis or a first degree family member with the disease.²

GI involvement can be seen in up to 25% of patients with NF, especially localized to the stomach and duodenum.³ Involvement of the colon is exceedingly rare. It is not common for neurofibromas to develop in a person who

does not have the diagnosis of NF1. This case report involves a 50 year old hispanic female with an isolated neurofibroma of the perianal region in the absence of any medical or family history of NF1.

Since there are very few cases reported in literature, it is important to document such cases and follow patients with this disease closely given that it can be an initial presentation of NF1 and because of its malignant potential.

CASE REPORT

A 50 year old hispanic female presented to the office with a perianal mass for 1 year that worsened in the last 2 weeks and was growing in size.

Patient denied rectal pain but admitted discomfort with bowel movements. Patient denied rectal bleeding, rectal discharge, fever, abdominal pain, nausea or vomiting. Patient had colonoscopy 1 week prior to visit with normal

findings: no polyps, ulcers or external masses were found.

Patient admits weight loss 15 pounds in 4 months. Her past medical history was significant for hypertension, hyperlipidemia, diabetes mellitus and uterine fibroids with a previous myomectomy. Family history significant for colon cancer in cousin, thyroid cancer in sister, breast cancer in mother.

Patient was hemodynamically stable when examined. On physical exam: No presence of Lisch Nodules, Abdomen was soft, nontender, nondistended with positive bowel sounds. There was no evidence of other skin tumors. Rectal exam was positive for perianal rubbery mass at 5 o'clock, measuring 2x2 cm non tender no active bleeding or hemorrhoids, good sphincter tone, Guaiac was negative.

The patient's labs were mostly noncontributory, with no evidence of anemia, with a hemoglobin of 14.1 and a hematocrit of 43.8, or infection, with a white blood cell count of 7.5 with no shifts. Patient was subsequently taken to OR for excision of the mass.

Surgical Intervention

During the procedure, a perianal mass was appreciated at 5 o'clock in lithotomy position. After skin incision, a pedunculated fibrous mass was identified, and excised measuring 5 x 3.3 x 1.4 cm. Mass was around external sphincter (Figure 1 A and B).

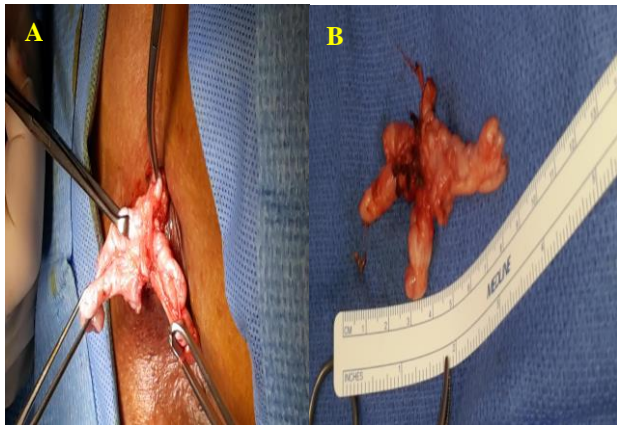


Figure 1: A) Intraoperative findings: a pedunculated fibrous mass was identified; B) excised measuring 5 x 3.3 x 1.4 cm mass was around the external anal sphincter.

Incision was closed primarily with chromic sutures. Pathology reported multilobulated fragment of tan-purple, rubbery, fibrous tissue, measuring 5 x 3.3 x 1.4 cm in greatest dimension and consistent with an anal tumor: Perianal Neurofibroma (Figure 2 A and B). Postoperatively patient did well and did not have any complaints. The incision healed with no complications.

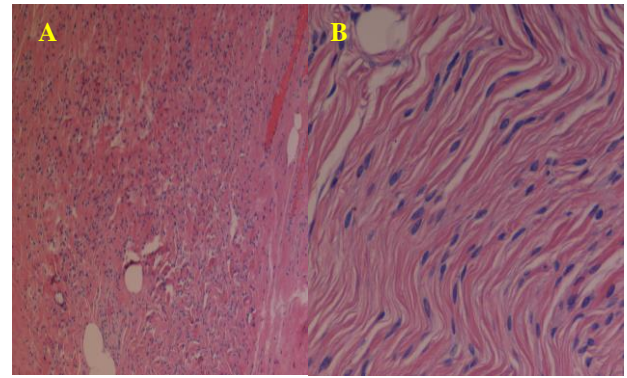


Figure 2: The specimen: consists (sent in formalin) of a multilobulated fragment of tan-purple, rubbery, fibrous tissue, measuring 5 x 3.3 x 1.4 cm in greatest dimension. microscopically: the abnormal cells reside in a matrix of loose, unorganized collagen fibers although arrangement into arrays can occur A) spindle shaped cells with elongated, wavy nuclei with scattered lymphocytes and mast cells; B) a herring bone appearance is characteristic of the nerve tissue tumors. diagnosis: perianal neurofibroma.

DISCUSSION

This patient had a rare instance of a solitary neurofibroma in the absence of other manifestations of Neurofibromatosis type 1 (NF1). NF1, as discussed, is an autosomal dominant genetic disorder from chromosome 17 that is characterized by multiple neurofibromas, lisch nodules, cafe au lait spots, optic gliomas and other features.⁴ Those without the diagnosis are generally not susceptible to developing neurofibromas due to the lack of the NF1 mutation. Neurofibromas can cause pain especially if located in the lumbosacral region and around the external anal sphincter, though GI involvement is uncommon.⁵ Any anorectal mass that causes pain must be carefully analyzed to rule out other possibilities. The differential diagnosis of an anorectal mass includes polyps, hemorrhoids, lymphogranuloma venereum, anorectal carcinoma, malignant melanoma and GIST tumor. Malignant tumors, such as anorectal malignant melanoma, that often carries a poor prognosis should be ruled out. Benign anal diseases include hemorrhoids, anal fissures, pruritus ani, perianal abscess and fistulas.⁶ Patients may present with perianal mass, discomfort, pain, discharge. These symptoms may mimic both malignant or benign masses of the anorectal region. It is important to consider the possibility of both benign or malignant neurogenic tumors of the perianal region and to rule these conditions to come to an accurate diagnosis with appropriate management.

Neurogenic tumors of the anorectal region include neurilemmomas and Neurofibromas. Both of these tumors are benign tumors that originate from the Schwann Cells of the peripheral nervous system.⁷ Neurofibromas can occur in the complex of an underlying background of NF1 which has the risk of malignant

transformation. It can also occur as a solitary nodule in the absence of a history of NF1. Surgical intervention is the current standard of care for symptomatic neurofibromas. Once excised, the mass will appear firm, pale gray, and homogenous on gross appearance and translucent when cut. Microscopically, tumor cells are spindle shaped with elongated, wavy nuclei with scattered lymphocytes and mast cells. The cells reside in a matrix of loose, unorganized collagen fibers although arrangement into arrays can occur. A herring bone appearance is characteristic of the nerve tissue tumors.⁸ Patients may present with mild symptoms and mimic hemorrhoids. Without intervention, neurofibroma may continue to grow in size, at which point patient may become symptomatic. Larger neurofibromas may lead to compression and displacement of critical organs, in the same way as a malignancy would.⁹ A surgical intervention and excision of the mass is both diagnostic and curative. However, due to presence of malformed vessels within the neurofibroma, there is an increased risk of bleeding.¹⁰ Careful excision with appropriate cautery and ligation to achieve hemostasis are adequate to prevent excessive bleeding. Solitary neurofibromas, such as the one in this patient, can be managed especially if the mass is detected early and removed with minimal complications.

CONCLUSION

In a patient presenting with a solitary perianal mass, it is important to consider the possibility of a neurofibroma. A solitary perirectal Neurofibroma may be the presenting symptom of Neurofibromatosis Type 1 which has malignant potential. Surgical intervention in such cases can be both diagnostic and curative.

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