Case Report
Middle East Journal of Cancer 2011; 2(1): 43-47

Large Neurofibroma of the Anorectal Canal: A Case Report

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Abstract

Neurofibromatosis type 1, also known as von Recklinghausen disease, is an autosomal dominant disorder with an incidence of 1 per 4000. Neurofibromas are benign, heterogeneous tumors arising from the connective tissue of peripheral nerve sheaths, especially the endoneurium. Visceral involvement in disseminated neurofibromatosis is rare. Neurofibroma occurs most frequently in the stomach and jejunum, but the colon and anorectal canal may also be involved. Gastrointestinal neurofibromas may lead to bleeding, obstruction, intussusception, protein-losing enteropathy and bowel perforation. We present a case of diffuse involvement of the anorectal area by a huge neurofibroma, with pelvic pain, watery diarrhea and urgency.

Keywords: Neurofibroma, Anorectal mass, Diarrhea

Introduction

Neurofibromatosis (NF) is an autosomal dominant disorder affecting approximately 1 in 4000 people among all ethnic groups.[1-3] The National Institute of Health Consensus Development Conference has defined two distinct types: neurofibromatosis type 1 (NF1) or von Recklinghausen disease, which affects 85% of patients, and neurofibroma type 2 or bilateral acoustic neuromas and vestibular schwannomas, which affect 15% of patients.[1] The diagnosis of NF1 is currently based on clinical criteria that include the presence of multiple skin lesions (café-au-lait), neurofibromal tumors, multiple freckles, osseous lesions, optic glioma, iris hamartoma, visceral involvement and other features.[1,3-5]

We report a young woman who presented with pelvic pain, watery diarrhea and urgency. Physical examination and work-up revealed multiple café-au-lait lesions and a large anorectal neurofibroma. Decreasing rectal capacity due to the large tumor caused fecal urgency. Extension of the tumor to the inter-sphincteric plane caused pelvic and perineal pain, but no fecal incontinence.

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Middle East Journal of Cancer

Recived: September 30, 2010; Accepted: October 18, 2010

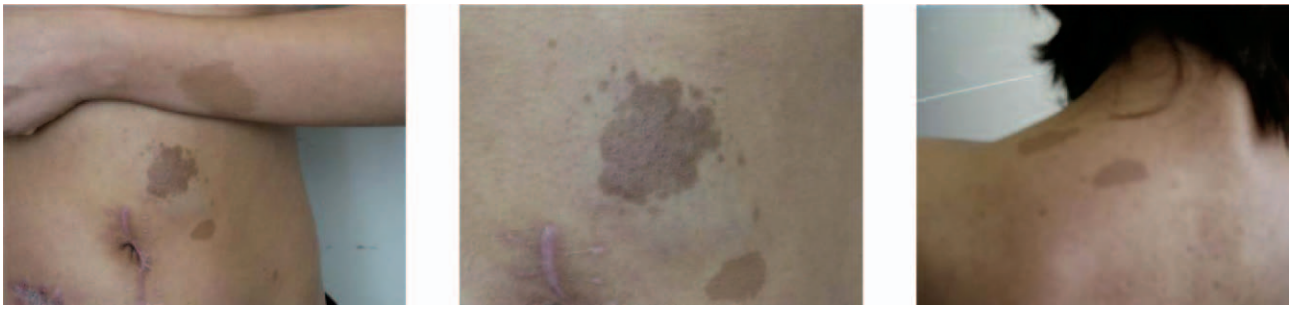

Figure 1. Café-au-lait skin lesions of variable size in the patient.

Case report

A 19-year-old woman presented two years before this writing with a 6-month history of pelvic pain, watery diarrhea and urgency without rectal bleeding. She had a sensation of incomplete evacuation and constant dull pelvic pain. She denied any previous medical problems. Her family history was unremarkable with the exception of similar multiple hyperpigmented skin lesions in her father, brother and sisters, who otherwise had no medical problems. Her personal and drug histories were normal.

General physical examination revealed a young woman with five café-au-lait skin hyperpigmentations of variable sizes (8 mm to 6 cm) that involved the trunk, abdomen and left forearm (Figure 1).

Digital rectal exam detected a huge circumferential extramural mass in the anorectal canal. The remainder of the physical examination was normal, particularly the abdomen. Laboratory studies including complete blood count, liver function test, serum chemistry, urinalysis and stool examination were normal. Endoanal sonography (B&K machine, 360-degree probe) showed a hypoechoic density around the anorectal canal and lymphadenopathy with size <10mm (Figure 2).

Colonoscopy showed a large, circumferential, submucosal mass in the anorectal canal that extended to the rectosigmoid junction, therefore a biopsy was taken. Pathologic analysis of the specimen revealed a benign spindle cell and herringbone appearance that favored a diagnosis of neurofibroma. The pelvic computed tomography scan with intravenous contrast revealed a large anorectal mass of about 15 cm that involved the perirectal fossa (Figure 3). Her anal manometry result was normal.

The patient underwent a transabdominal pelvic exploration via a low midline incision. During surgery, multiple adhesions of the small and large bowel to the pelvic organs were released. A firm, fleshy, whitish tumor was attached to the intra- and extraperitoneal parts of the rectum with extension to the anal canal (Figure 4). Total mesorectal

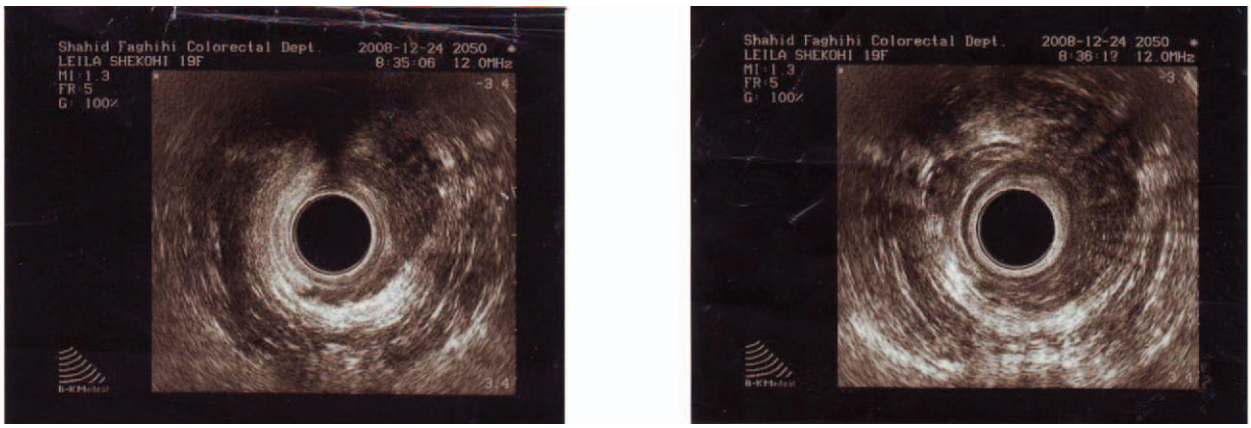

Figure 2. Endoanal sonography showing a hypoechoic density around the anorectal canal.

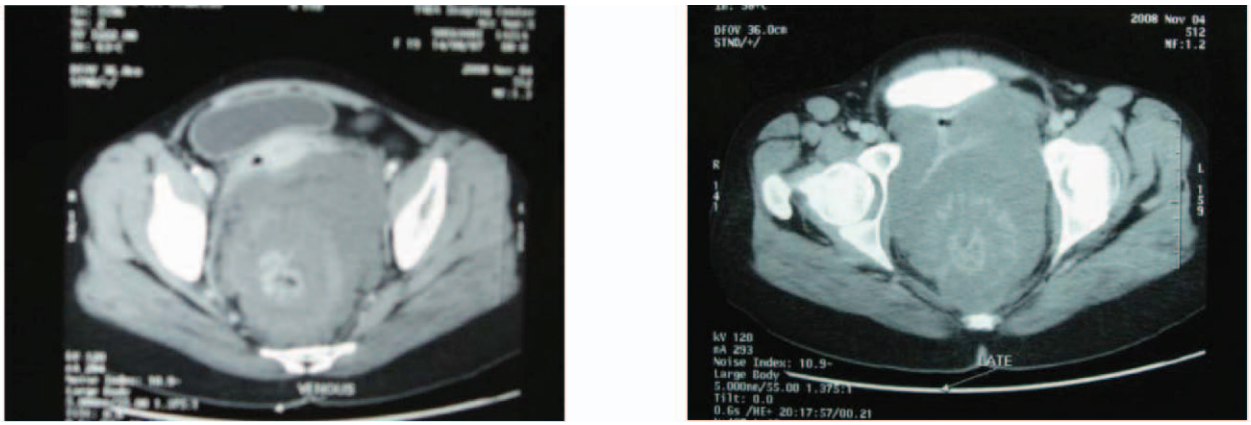

Figure 3. Pelvic computed tomography scan with intravenous contrast showing a large anorectal mass that involved the perirectal fossa.

excision with colo–anal anastomosis and diverting ileostomy were performed. A number 31 circular stapler was used for colo–anal anastomosis.

Pathologic examination revealed a 12 × 9 × 3 cm creamy white mass that occupied the entire circumference of the rectum and anal canal with extension to the mucosa and serosa, without malignant changes (Figure 5). Immunohistochemical tests including CD34 (QBEnd 10, Dako-Denmark), C-Kit (Polyclonal, Dako- Denmark) were negative, whereas S-100 stain (QBEnd 10, Dako- Denmark) was positive, a finding that favored a diagnosis of neurofibroma.

Discussion

Neurofibromatosis type 1 is a multisystemic disorder that may affect any organ in the body.[6] The NF1 gene has been identified on chromosome 17q11.2.[7] Of the nerve sheath tumors, neurofibromas occur more frequently than schwannomas and tend to affect younger patients without gender preference.[8] When present in deeper soft tissues and viscera, neurofibromas may become quite large and seem to be encapsulated.[9]

Visceral involvement in disseminated NF is rare. Gastrointestinal involvement in NF occurs in as many as 25% of cases,[10] whereas only about 15% of the cases with NF in the gastrointestinal (GI) tract are associated with von Recklinghausen disease.[11] As the tumor enlarges, the overlying mucosa becomes ulcerated and bleeds. Intussusception, obstruction and bowel perforation are recognized complications.[12] Patients may present with abdominal pain, nausea, abdominal distention, diarrhea, constipation, bowel perforation and GI bleeding.[13] Most previously reported cases have described lower GI bleeding in NF1 with colonic involvement by a gastrointestinal stromal tumor (GIST).[14] It is known that patients with NF1 are at increased risk for developing both benign and malignant tumors,

Figure 4. Gross aspect of the tumor removed during transabdominal pelvic exploration.

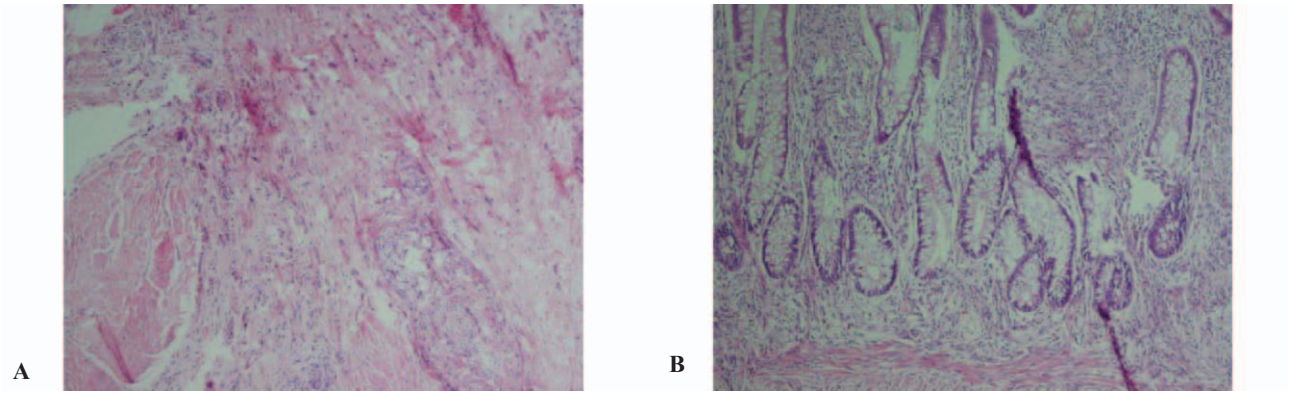

Figure 5. Histopathologicalexamination of the tumor showed no evidence of malignant change. A, H.E stain, 40×. B, H.E stain, 10×.

predominantly derived from the neural crest.[15] Neurofibromas in NF1 may undergo malignant change (sarcomatous degeneration) in 3% to 15% of patients.[12, 13] Differential diagnoses of the anal mass include polyps, hemorrhoids, lymphogranuloma venereum, anorectal carcinoma, malignant melanoma and GIST tumor.[9]

Gastrointestinal neurofibromas may be diffuse and submucosal, thus radiographic visualization is difficult. Imaging plays an important role in the diagnosis, evaluation and follow-up of patients with abdominal manifestations of NF1. Endoanal sonography can reveal a hypoechoic, lobulated tumor with smooth, well-defined margins. Barium studies may demonstrate an extraluminal mass effect. Computed tomography scan may reveal a solid mass with central areas of low attenuation and occasional calcification. The mass is usually well defined and has homogenous low attenuation equal to or slightly higher than water, but lower than muscle. Magnetic resonance imaging is considered the modality of choice.[6, 16] However, due to its rarity, a high degree of suspicion is needed for a timely diagnosis. Delays in the diagnosis of GI neurofibroma are common.[6]

Surgical resection is the treatment of choice for all symptomatic tumors in patients with NF1.[6] Grossly, neurofibromas are firm, pale gray, homogenous and translucent when cut. Microscopically, tumor cells are spindle-shaped with elongated, wavy nuclei. Scattered among these cells are lymphocytes and mast cells. The cells reside in a matrix of loose, unorganized collagen fibers although arrangement into arrays can occur. A herringbone appearance is characteristic of nerve tissue tumors.[8,12] Malignant GIST with multiple metastases can be treated with sunitinib, as previously reported in a patient with NF1.[17]

A large neurofibroma of the anorectal canal is rare. We initially evaluated the patient described here for other manifestations of NF1. All work-ups were negative with the exception of digital rectal examination, which disclosed a huge mass in the anorectal canal. Colonoscopic biopsy revealed a neurofibroma and resection was suggested. The large neurofibroma of the rectum extended to the intersphincteric space of the anal canal and caused pelvic pain, urgency and watery diarrhea, but no lower GI bleeding or fecal incontinence. The size and extent of this tumor were features of note for us in such a young person.

In conclusion, consideration of neurofibroma in the differential diagnosis of an anal mass, particularly in patients with NF1, is important because resection alone is the treatment of choice.

References

1. Huson SM, Hughen RAC. The neurofibromatosis: A pathogenetic and clinical overview. London: Chapman and Hall, 1994.
2. Riccardi VM, Eichner JE. Neurofibromatosis: Phenotype natural history and pathogenesis. Baltimore: Johns Hopkins University Press, 1986.
3. Ishii S, Han S, Shiiba K, Mizoi T, Okabe M, Hoii A, et al. Allelic loss of the NF1 gene in anal malignant melanoma in a patient with neurofibromatosis type 1. *Int J Clin Oncol* 2001(August); 6(4): 201-4.
4. Gutmann DH, Aylswerth A, Carey JC, Korf B, Marks J, Pyeritz RE, et al. The diagnostic evaluation and

multi-disciplinary management of neurofibromatosis 1 and 2. *JAMA*. 1997;278:51-7.
5. National Institutes of Health Consensus Development Conference. *Arch Neural* 1988; 45:575-8.
6. Rastogi R. Intra-abdominal manifestation of von Recklinghausen's neurofibromatosis. *Saudi Journal of Gastroenterology*. 2008;14:80-2.
7. Ferner R. E, O'Doherty M. J. Neurofibroma and schwannoma. *Current Opinion in Neurology*. 2002;15:679-84.
8. James E, Linda J, Vargas HD. Solitary neurofibroma of the anal canal, report of two cases. *Dis Colon Rectum* 2000;43:109-12.
9. Harkin J, Reed R. Tumors of the peripheral nervous system. In: Atlas of tumor pathology. Mishawaka, IN, U.S.A. *Armed Forces Institute of Pathology*, 1969;29-84.
10. Abdonabi S. Alkaraphouli Y, Abdulla S. Neurofibromatosis with lower GIT presentation, case report. *IJGE* 2006;1: 60-2.
11. Samuel I, Jakate S, Ramsey M, Saclarides TJ. Abdominal mass in a 19-year-old with neurofibromatosis. *Postgrad Med J* 1997;73:325-6.
12. Corman ML, Brown B, McMullan E, Gunning F, Rivera B, Glazer A. Less common tumors and tumorlike lesion of the colon, rectum and anus. In: Colon and rectal surgery. New York . Publisher: Lippincott Williams & Wilkins, 2005;1123-5.
13. Nawazkhan A, Turnbull I, MacDonald S, Desai N, Bibi N, Al-Okaili R. Neurofibromatosis type 1. Medicine Radiology. *Medscape.com*. Updated; Jan 2008. Available at http://emedicine.medscape.com /article/342500-overview.
14. Pinsk I, Dukhno O, Ovant A, Levy I. Gastrointestinal complications of von Recklinghausen's disease: Two case reports and a review of the literature. *Scand J Gastroenterol* 2003;38(12): 1275-78.
15. Seiichi I, Shunagyin H, Ken IS. Allelic loss of the NFI gene in anal malignant melanoma in a patient with neurofibromatosis type 1. *Int J Clin Oncol* 2001;6(4): 201-4.
16. Zacharia T, Jaramillo D, Poussaint T, Korf B. MR imaging of abdominopelvic involvement in neurofibromatosis. Type 1: A review of 43 patients. *Pediatr Radiol* 2005;35:317-22.
17. Kalender M, Sevinc A, Tutar E, Sirikci A, Camci C. Effect of sunitinib on metastatic gastrointestinal stromal tumor in patients with neurofibromatosis type 1: A case report. *World J Gastroenterol* 2007;13(18):2629-32.