Neurofibroma of the anorectal canal presenting with diarrhoea and pelvic pain: Report a Case

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Abstract

Neurofibromatosis type-1 (NF1), also known as Von Recklinghausen’s disease, is an autosomal dominant disorder with incidence of one in 4,000. Neurofibromas are benign, heterogeneous, peripheral nerve sheath tumours arising from the connective tissue of peripheral nerve sheaths, especially the endometrium. Visceral involvement in disseminated neurofibromatosis is considered rare. Neurofibroma occurs most frequently in the stomach and jejunum, but the colon and anorectal canal may be also involved. Gastrointestinal neurofibromas may lead to bleeding, obstruction, intussusception, protein-losing enteropathy and bowel perforation. We encountered a difficult case with a huge neurofibroma involving the anorectal area. The patient presented with pelvic pain, watery diarrhoea and urgency.

KEYWORDS: neurofibroma, anorectal mass, diarrhoea
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Abstract:

Neurofibromatosis type-1 (NF1), also known as Von Recklinghausen’s disease, is an autosomal dominant disorder with incidence of one in 4,000. Neurofibromas are benign, heterogeneous, peripheral nerve sheath tumours arising from the connective tissue of peripheral nerve sheaths, especially the endometrium. Visceral involvement in disseminated neurofibromatosis is considered rare. Neurofibroma occurs most frequently in the stomach and jejunum, but the colon and anorectal canal may be also involved. Gastrointestinal neurofibromas may lead to bleeding, obstruction, intussusception, protein-losing enteropathy and bowel perforation. We encountered a difficult case with a huge neurofibroma involving the anorectal area. The patient presented with pelvic pain, watery diarrhoea and urgency.

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Introduction:

Neurofibromatosis (NF) is an autosomal dominant disorder affecting approximately 1 in 4000 people in all ethnic groups. (1-3) The National Institute of Health (NIH) consensus development conference has defined two distinct types: neurofibromatosis type 1 (NF1) or Von Recklinghausen’s disease, which affects 85-90% of patients and neurofibroma type 2 (NF2) or bilateral acoustic neuromas/vestibular Schwannomas which affects 10% of patients. (1)

The diagnosis of NF1 is currently based on clinical criteria including the presence of multiple skin lesions (café-au-lait spots), neurofibroma tumours, multiple freckles, osseous lesions, optic glioma, iris hamartoma, visceral involvement and other features.(1,3-8)

In this rare case we report a young female patient who presented with pelvic pain, watery diarrhoea and urgency. Her physical examination revealed multiple café-au-lait skin lesions and digital examination revealed a large mass.
Case report:

In 2007 a 19-year-old female presented with a history of pelvic pain, watery diarrhoea and urgency but no history of rectal bleeding. She had a sensation of incomplete evacuation and a constant dull pelvic pain. She denied any previous medical problems. However, her family history indicated that her father, brother and sisters all have similar multiple hyperpigmental skin lesions.

General physical examination revealed a young female with five café-au-lait skin hyperpigmentations of varying sizes (8mm-6cm) involving the trunk, abdomen and left forearm (figures 1, 2).

A large circumferential extramural mass in the anorectal canal was detected by digital rectal examination. The remainder of physical examination was normal and laboratory investigations including complete Blood Count, Liver Function Tests and Serum chemistry were also normal. Endoanal sonography showed hypoechoic density around anorectal canal (figure 3).

Colonoscopy showed a large circumferential submucosal mass in the anorectal canal extending to the rectosigmoid junction which was biopsied. Histopathology of the biopsy revealed a benign spindle cell and herringbone appearance suggestive of neurofibroma. A subsequent pelvic CT scan with IV contrast revealed a large anorectal mass about 15cm long involving the anorectum (figure4). Her anal manometry was normal.

The patient was scheduled for a laparoscopic operation but due to the presence of dense small and large adhesions to the pelvic organs the procedure was converted to open. A firm, fleshy, whitish tumour attached to the intraperitoneal and extraperitoneal part of the rectum and anal canal was resected. Total mesorectal excision was undertaken together with a coloanal anastomosis performed using circular stapler number 31. A diverting ileostomy was established.
A 12x9x3cm creamy white mass which occupied the whole circumference of rectum and anal canal was sent to pathology (figure 5). Pathology revealed that the mass extended to the mucosa and serosa but confirmed there were no malignant changes (Figure 6, 7). Immunohistochemistry including CD-34, C-kit were negative, but S-100 was positive in favour of neurofibroma.
Discussion:

Neurofibromatosis type-1 is a multisystemic disorder that may affect any organ in the body. (9) The NF1 gene was identified on chromosome 17q11.2 (10) of the nerve sheath tumours and neurofibromas occur more frequently than Schwannomas. They tend to affect younger patients and do not indicate gender preference (14). When occurring in deeper soft tissues and viscera, neurofibromas may become quite large and appear to be encapsulated (13).

Visceral involvement in disseminated neurofibromatosis is considered rare however gastrointestinal involvement of neurofibromatosis occur in as many as 25% of cases (11). While only about 15% of these are associated with Von Recklinghausen’s disease (12). As the tumour enlarges, the overlying mucosa becomes ulcerated and bleeds. Intussusception, obstruction and bowel perforation are recognized complications (7). Patients may present with abdominal pain, nausea, abdominal distension, diarrhoea, constipation, bowel perforation and GI bleeding. (8) Cameron et al noted that severe diarrhoea sometimes occurred as a symptom among patients with Von Recklinghausen’s neurofibromatosis as a concomitant occurrence with an adrenal ganglioneuroma and a pheochromocytoma (13). In our case the CT scan did not show either.

In addition to neurofibroma, it is known that patients with NF1 are at increased risk of the development of both benign and malignant tumours predominantly derived from neural crest (3). Neurofibromas in NF1 may undergo malignant changes (sarcomatosis degeneration) in 3-15% of patients (7, 8). The differential diagnoses of anal masses include polyps, haemorrhoids, lympho-granuloma veneroums, anorectal carcinomas, malignant melanoma and GIST tumours must also be considered (14).
When the gastrointestinal neurofibroma is diffuse and submucosal the radiological appearance is difficult to interpret although imaging is important in the diagnosis, evaluation and follow-up of patients with abdominal manifestation of NF1. Endoanal sonography shows hypoechoic, lobulated but smooth and well-defined margins. Barium studies may demonstrate extra luminal mass effect. CT scans 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 more than water, but lower than muscle. MRIs are considered to be the modality of choice (9, 15). But due to its rarity the diagnosis of GI neurofibroma is often delayed, usually only being made following tests for other, more common occurring, conditions (9).

Surgical resection is the preferred treatment for all symptomatic tumours occurring in patients with NF1(9). Macroscopically neurofibroma is firm, pale gray, homogenous and translucent when sectioned. Microscopically, tumour cells are spindle-shaped with elongated and wavy nuclei. Scattered among these cells are lymphocytes and mast cells. The cells reside in a matrix of unorganized and loose collagen fibre although arrangement into arrays can occur. Herringbone appearance is characteristic of nerve tissue tumour (7, 16). Diarrhoea is a rare symptom of neurofibroma and after surgery the diarrhoea resolved completely for this patient.

A large neurofibroma lesion of the anorectal region is rare. In our case, our initial diagnosis was of a GIST tumour only but further physical examination and the result of the biopsies via colonoscopy allowed the diagnosis of neurofibroma to be established.

Conclusion

Consideration of neurofibroma as the differential diagnosis of an anal mass especially NF1 is important because resection alone is the treatment of choice.
References:


Figures

**Figures 1, 2:** Hyperpigmentation

**Figure 3:** Endorectal ultrasound showing extramural mass

**Figure 4:** Pelvic CT scan showing large anorectal mass

**Figure 5:** Resected specimen
Figures 6 and 7: Pathology slides showing neurufibroma of the colorectal; Bland fusiform tumor cells between rectal mucosal glands in wormlike Growth pattern. (Original magnification x 40)