

Solitary colonic neurofibroma in an African child

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Neurofibromas are a group of heterogeneous neurocutaneous disorders. They are benign neoplasms consisting of neural and connective tissue components affecting any organ system. Gastrointestinal involvement in neurofibromatosis type 1 (NF1) are rare and are late manifestations of the disease, but in exceptional cases they can be the initial sign of neurofibromatosis in patients who have no external stigmata that arouse suspicion. Neurofibromatosis of the colon as a part of the NF1 is rare. Isolated colonic neurofibromatosis without other features suggestive of NF1 is rarely encountered in clinical practice. We report the case of a 12-year-old boy with an isolated colonic neurofibromatosis presenting with a right hypochondrial mass with no external features of NF1.

Introduction

Neurofibromas are benign tumours that may grow on nerves anywhere in the body. An autosomal dominant disorder, neurofibromatosis affects any organ system including the gastrointestinal tract [1]. Neurofibromatosis of the stomach and small intestine are rare and associated with von Recklinghausen's disease also known as neurofibromatosis type 1 (NF1) [2]. Rarer still is isolated neurofibromatosis of the colon [3] and its occurrence in the paediatric age group. We report an isolated neurofibromatosis of the colon presenting with no systemic symptoms in an African child.

Case report

A 12-year-old boy presented with 3-month history of painless, left hypochondrial abdominal mass. There was no weight loss, jaundice, constipation, passage of bloody watery stool and no family history of neurofibromatosis. Examination revealed a nontender and firm left hypochondrial mass. There were no café-au-lait spots or Lisch nodules and no other cutaneous lesions. Abdominal ultrasonography revealed splenomegaly and a pear-shaped mass measuring 8.5 cm × 4.9 cm, solid with calcific specks inferomedial and separate from the spleen. Fine needle aspiration cytology of the mass revealed a smear of fibromyxoid stroma with spindle cells.

He underwent an exploratory laparotomy, which revealed hard, round, splenic flexure colonic mass, almost occluding the lumen and adherent to both the spleen and greater curvature of the stomach. Splenectomy was performed and the colonic mass resected with an end-to-end anastomosis. He was discharged home following an uneventful postoperative recovery.

Histological sections of the mass showed a benign mesenchymal neoplasm composed of proliferating Schwann cells and fibroblasts dispersed in a loose myxoid stroma; the cells have slender, spindle-shaped, wavy nuclei and moderate eosinophilic cytoplasm, thus suggesting neurofibroma of the colon (Fig. 1).

We report this case as its presentation in children may give a diagnostic dilemma and the probability of malignant digestive disease associated with NF1 should be kept in mind, regardless of the age of the patient. *Ann Pediatr Surg* 14:87–88 © 2018 Annals of Pediatric Surgery.

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Discussion

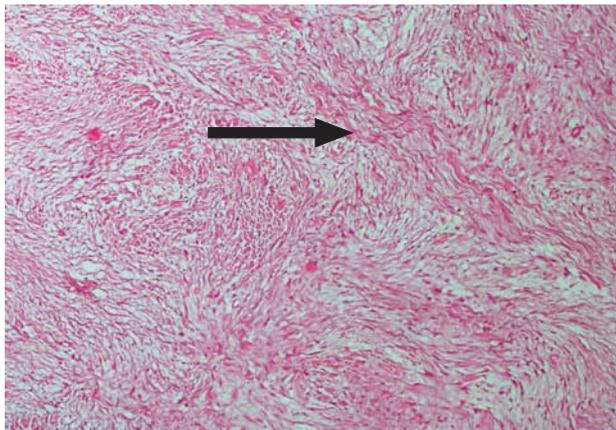
Neurofibromas are a group of heterogeneous neurocutaneous disorders. They are benign neoplasms consisting of neural and connective tissue components. Neurofibromatosis are usually manifestations of the von Recklinghausen's disease (NF1) consisting of Schwann, perineural cells and myofibroblasts [3].

Neurofibromas of the digestive system are usually found as a part of NF1 [3] and commonly found in the stomach, duodenum, jejunum and ileum as reported in 25% of all cases, but any portion of the gastrointestinal tract may be involved [3,4]. Gastrointestinal neurofibromas are usually late manifestations of the disease, but in exceptional cases they can be the initial sign of neurofibromatosis in patients who have no external stigmata that arouse suspicion. The occurrence of gastrointestinal neurofibromas should, therefore, lead to a careful search for other features of NF1 in affected patients and their families [5].

Colonic neurofibromatosis as a part of NF1 is rare [3,5]; rarer still is isolated colonic neurofibromatosis without other features suggestive of NF1 [3]. It may be familial or sporadic in presentation, whereas the majority may be associated with NF1. The tumours are frequently sessile and wide based with associated pedunculated polyps [3,6].

Clinically, it may remain silent and rarely cause symptoms before puberty [3]. Presentation may include constipation or diarrhoea [7], bleeding from mucosal necrosis or ulceration [2], palpable abdominal masses [8], abdominal pain [9] and intestinal obstruction [10]. Colonic lesion may present with myriad of symptoms, which often depends on its location, whether lesion is diffuse or focal in nature, its effect on gastrointestinal motility, and possible pressure effects on adjacent structures [11]. Solitary intestinal lesions may manifest with intestinal symptoms without any demonstrable clinical evidence of associated systemic syndromes [11] as seen in the case presented, whereas the classic features of café-au-lait

Fig. 1



Histopathological image of the colonic neurofibroma showing spindle-shaped Schwann cells having elongated wavy nuclei and eosinophilic cytoplasm (arrow) and fibroblastic cells dispersed in a loose myxoid stroma. Haematoxylin and eosin stain, $\times 100$.

spots, Lisch nodules and cutaneous neurofibromas may be seen in patients with NF1.

Delay in the diagnosis of isolated gastrointestinal neurofibromatosis is common due to its rare nature; the much rarer isolated colonic neurofibroma is even more difficult to diagnose. Only 13 cases of isolated colonic neurofibromatosis with no evidence of NF1 have been documented in the literature [3] with two reports of isolated colonic neurofibromas, one of which only presented with a palpable mass involving the transverse colon similar to our patient's presentation.

This type of presentation often gives diagnostic dilemma in affected children; thus, the possibility of malignant digestive disease associated with NF1 should be entertained in

patients, regardless of age and neurofibromatosis characteristics [4]. Therefore, a high index of suspicion is needed in making the diagnosis, especially when they present with an abdominal mass with no pressure effects on adjacent structures.

Conflicts of interest

There are no conflicts of interest.

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