

Solitary plexiform neurofibroma of the stomach: A case report

Lei Shi, Fu-Jian Liu, Qiu-Hong Jia, Hang Guan, Zhao-Jiong Lu

Lei Shi, Fu-Jian Liu, Qiu-Hong Jia, Hang Guan, Zhao-Jiong Lu, Department of Gastroenterology, the Eighth Affiliated Hospital of Guangxi Medical University, Guigang 537100, Guangxi Zhuang Autonomous Region, China

Author contributions: Shi L conceived and coordinated the study, and participated in data collection and manuscript writing; Liu FJ participated in the study design, data collection, production of histopathological figures, and manuscript writing; Jia QH, Guan H and Lu ZJ participated in data collection and manuscript writing.

Correspondence to: Lei Shi, MD, PhD, Department of Gastroenterology, the Eighth Affiliated Hospital of Guangxi Medical University, No. 1 Zhongshan Road, Guigang 537100, Guangxi Zhuang Autonomous Region, China. ycymoment@hotmail.com
Telephone: +86-775-4200028 Fax: +86-775-4200028

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Abstract

Plexiform neurofibroma (PN) of the digestive tract is very rare and usually part of the generalized syndrome of neurofibromatosis type 1 (von Recklinghausen disease). Solitary PN of the stomach is extremely rare and has not been reported in the literatures. Here we present a case of solitary PN of the stomach, which was not associated with von Recklinghausen disease. A 38-year-old male presented abdominal pain and distention for 7 d. The patient underwent endoscopy of the upper gastrointestinal tract, which revealed a 3.5 cm protruding and cauliflower-shaped mass with a shallow 1 cm central ulcer in the greater curvature of the stomach. The lesion was removed by laparoscopic surgery. Histological examination demonstrated characteristic histological findings of spindle-shaped cells. Immunohistochemical analysis showed that the tumor cells were positive for S-100 protein, but negative for CD34, KI-67, CD117, and actin. Based on histological findings, gastrointestinal stromal tumor could be excluded, and thus the case was confirmed as PN. We described the

clinical features, physical examination, endoscopic findings, and histopathological examination of this case.

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Key words: Plexiform neurofibroma; Neurofibromatosis; Von Recklinghausen disease; Abdominal pain

Core tip: Solitary plexiform neurofibroma (PN) is extremely rare. We report a case of solitary PN of the stomach in a 38-year-old male who underwent laparoscopic surgery. The case was not associated with von Recklinghausen disease, and the patient was in good condition 6 mo after surgery, with no tumor recurrence. To our knowledge, this is the first reported case of isolated stomach PN undergoing laparoscopic surgery. Endoscopic treatment is technically feasible and may be considered as the procedure of choice for solitary PN treatment. A long-term follow-up endoscopy of the upper gastrointestinal tract is greatly needed.

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INTRODUCTION

Plexiform neurofibroma (PN) is a rare benign peripheral nerve sheath tumor mainly associated with von Recklinghausen disease. PN is rarely presented as a sporadic lesion in a patient without features of generalized neurofibromatosis^[1]. Up until now, there were no reports of solitary PN of the stomach without evidence of neurofibromatosis type 1 (NF1) in the literature. Here we describe a rare case of a solitary PN of the stomach in a patient without

features of NF1.

CASE REPORT

A 38-year-old man was admitted to our department due to abdominal pain and distention for 7 d. Informed consent was obtained from the patient. No other signs of von Recklinghausen disease were found in this patient or his family. Physical examination revealed moderate tenderness on palpation of the upper abdominal region. Physical examination of the lungs, heart, and other systems revealed no abnormal features. The patient had no significant medical background or family history. No abnormal findings were revealed by laboratory tests. Chest X-ray, electrocardiography, and abdominal sonogram showed no abnormal features. For diagnostic purposes, an endoscopy of the upper gastrointestinal tract was performed, which revealed a 3.5 cm protruding and cauliflower-shaped mass with a shallow 1 cm central ulcer in the greater curvature of the stomach (Figure 1). Further endoscopic examination revealed no lesions in the small bowel or colon. Computed tomography (CT) imaging with and without contrast medium revealed a soft tissue mass-like lesion about 2.5 cm × 3.5 cm in the greater curvature of the stomach (Figure 2). Upper gastrointestinal radiography showed a mass in the greater curvature of the stomach (Figure 3). The patient underwent an endoscopic submucosal dissection for the en-bloc resection of the lesion. Injection of saline into the submucosa during gastroscopy did not completely elevate the tumor. Therefore, local gastroscopic resection was not indicated. Laparoscopic surgery was successfully performed.

The tumor was diagnosed as sporadic PN, which rarely occurs in the digestive tract. The diagnosis was confirmed by laparoscopic surgery with complete resection. Histological examination demonstrated characteristic histological findings of spindle-shaped cells. Immunohistochemical analysis showed that the tumor cells were positive for S-100 protein (Figure 4), but negative for CD34, KI-67, CD117, and actin. Gastrointestinal stromal tumor (GIST) could thus be excluded in our case and confirmed as PN. Furthermore, we performed PCR and sequencing on a blood sample from the patient and found no any mutations in the *NF1* gene (data not shown). We performed upper gastrointestinal endoscopy six months post-operatively for follow-up, and found no evidence of recurrence.

DISCUSSION

Neurofibromatosis is a benign peripheral nerve sheath tumor. It is a neurocutaneous disorder with two clinical forms: (1) NF1, described by von Recklinghausen; and (2) central type 2, described as a central neurofibromatosis that mainly affects the central nervous system^[2]. NF1 is the most common type of neurofibromatosis and accounts for about 90% of all cases. An autosomal-dominant mutation of the neurofibromin (tumor suppressor)



Figure 1 Endoscopic examination revealed a 3.5 cm protruding and cauliflower-shaped mass with a shallow 1 cm central ulcer in the greater curvature of the stomach.

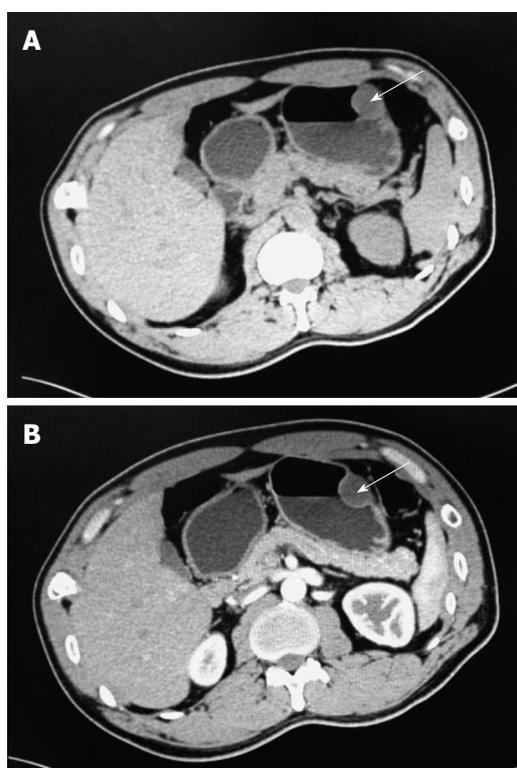


Figure 2 Radiological images of the abdomen. A: Computed tomography (CT) showed a soft tissue mass-like lesion about 2.5 cm × 3.5 cm in the greater curvature of the stomach (arrow); B: Post-contrast abdominal CT shows the same area not representing the contrast-enhanced appearance (arrow).

gene on chromosome 17 causes tumors of the peripheral nerves known as neurofibromas. NF2 is an autosomal-dominant disorder that induces tumor development. NF-1 is a systemic disorder that may affect any organ in the body, with clinical presentation depending upon the body system involved. Diagnosis of NF1 is based upon a series of clinical criteria as defined by the National Institutes of Health Consensus Statement^[3]: (1) at least six so-called “café-au-lait” spots in post-pubertal patients; (2) at least two neurofibromas of any type or one PN; (3) axillary or groin freckling; (4) at least two so-called “Lisch



Figure 3 Upper gastrointestinal radiography showed a mass in the greater curvature of the stomach (arrow).

nodules”; (5) optic glioma; (6) distinctive osseous bony dysplasia (such as thinning of the long bone cortex or sphenoid wing dysplasia; and (7) a close family member with NF1. The patient should have at least two of the previous criteria to be considered as NF1.

PN has a network-like growth involving multiple fascicles of a nerve, and may include multiple branches of a large nerve trunk. PN is a rare benign tumor developing from Schwann cells of the peripheral nervous system, often associated with NF1. Isolated PN in other locations without NF1 features has been very rarely encountered in clinical practice. The few cases have been reported so far have been in the oral cavity^[4], submandibular salivary gland^[1], lesser omentum^[5], penis^[6], cheek mucosa^[7], and intraparotid facial nerve^[8]. The specific cause and development of solitary PN are not known, but the possibility of mosaicism of NF-1 or a related gene has been considered. It is supposed that solitary PN may be caused by a sporadic localized mutation of the NF1 tumor suppressor gene^[9]. However, in this case, we found no direct evidence of NF1 mutation.

To our knowledge, this is the first report on the rare case of PN in the stomach. The 38-year-old male patient was diagnosed as PN based on clinical appearance, physical examination, and histological features. Our case should be distinguished from GIST, which usually occurs in the small intestine as multiple and clinically indolent tumors. GIST is consistently CD117 (KIT)-positive, generally CD34-positive, occasionally actin-positive, and rarely S-100-positive^[10]. According to immunohistochemical analysis, the tumor from this case was positive for S-100 protein, but negative for CD34, KI-67, CD117, and actin. Thus, GIST could be excluded in this case.

Although the cause and development of this rare case remain speculative, due to the malignancy potential, surgical resection, such as open surgery, combined endoscopic/laparoscopic intragastric resection, and laparoscopically gastroesophageal resection, is advocated^[11]. The pre-operative diagnosis of PN is often difficult. As a result, PN usually needs to be resected surgically. The application of fine needle aspiration (FNA), including computed tomography guided, transabdominal ultraso-

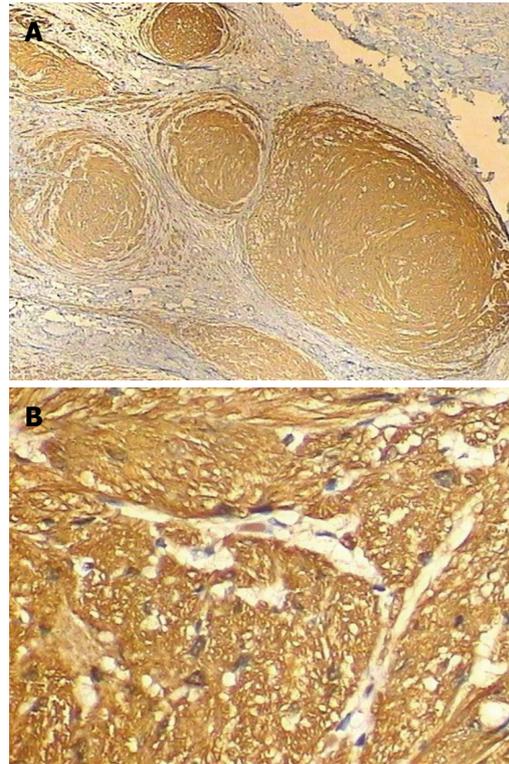


Figure 4 Histological examination of the gastric lesion. A: Histological findings showing irregularly distorted, enlarged nerve bundles ($\times 40$); B: Histopathological examination revealed nuclear and cytoplasmic reactivity with S-100 protein immunostain ($\times 400$).

nography guided, and endoscopic ultrasonography guided FNA, facilitates the diagnosis of GIST^[12]. However, FNA evaluation should not replace subsequent examination of resected specimens^[13].

With the wide application of novel techniques, including endoscopic mucosal resection and endoscopic submucosal dissection, endoscopic resection has recently been performed as a curative treatment for adenoma and early colorectal cancer^[14]. Endoscopic resection provides a better quality of life for the patient than conventional open surgery, as it is minimally invasive, brings complete resection, and supports detailed histopathologic evaluation of the specimens.

Although PN is generally benign, prompt resection is recommended because of a risk of malignant transformation. We advocated that endoscopic treatment is technically feasible and may be considered as the procedure of choice for solitary PN. Furthermore, a long-term follow-up endoscopy of the upper gastrointestinal tract is greatly needed.

COMMENTS

Case characteristics

A 38-year-old male was admitted to their department on an emergency basis because of abdominal pain and distention for 7 d.

Clinical diagnosis

Physical examination revealed moderate tenderness on palpation of the upper abdominal region.

Differential diagnosis

Immunohistochemical analysis showed that this case could be excluded as gastrointestinal stromal tumor.

Laboratory diagnosis

No abnormal findings were revealed by routine laboratory tests.

Imaging diagnosis

Computed tomography and upper gastrointestinal radiography showed a mass in the greater curvature of the stomach.

Pathological diagnosis

Histological examination demonstrated characteristic histological findings of spindle-shaped cells.

Treatment

Laparoscopic surgery was successfully performed.

Related reports

Isolated plexiform neurofibroma (PN) in other body organs without features of NF1 has been very rarely encountered in clinical practice. The few cases of isolated PN that have been reported so far have been located in the oral cavity, submandibular salivary gland, lesser omentum, penis, cheek mucosa, and intraparotid facial nerve.

Term explanation

Plexiform neurofibroma is a rare benign peripheral nerve sheath tumor mostly associated with von Recklinghausen disease and rarely presenting as a sporadic lesion in a patient without features of generalized neurofibromatosis.

Experiences and lessons

Immunohistochemical analysis should be performed to distinguish plexiform neurofibroma from gastrointestinal stromal tumors.

Peer review

This is the first report on a rare case of plexiform neurofibroma in the stomach. The authors described the clinical features, physical examination, endoscopic findings, and histopathological examination of this case. However, further information on the expression of the *NF1* gene and histological diagnosis will help us further understand the pathogenesis of this case.

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