CASE REPORT

Gangliocytic paraganglioma, a rare ampullary tumour treated with open transduodenal resection and sphincteroplasty

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SUMMARY

Gangliocytic paragangliomas are rare neuroendocrine tumours residing in the gastrointestinal tract, most commonly the periampullary region. Most are benign tumours with a low malignancy rate. We report a 50-year-old man who presented with acute onset of left-upper quadrant abdominal pain with radiation to the back. An intraluminal mass at the junction of the duodenum with normal pancreatic/hepatic parenchyma was discovered on abdominal CT. Following upper endoscopy and MRI revealing a periampullary lesion, fine-needle aspiration and biopsies were. Immunohistochemistry was positive for synaptophysin and S100HU, consistent with gangliocytic paraganglioma. The benign nature of this tumour and unique anatomy of a separate opening of the pancreatic and common bile ducts led to transduodenal excision with sphincteroplasty, thereby avoiding extensive surgery. Surveillance CT every 6 months and upper endoscopy initially every 6 months (now yearly) revealed no evidence of endoscopic or histological recurrence at 3 years follow-up.

BACKGROUND

Gangliocytic paragangliomas are rare neuroendocrine tumours mainly residing in the gastrointestinal tract, with the most common location being in the periampullary region. In most cases, it represents a benign tumour with a <5% malignant potential.¹ Dahlin and coworkers² first described this tumour in 1957, and Taylor and Helwig³ reported benign non-chromaffin paragangliomas of the duodenum in 1962. The term 'gangliocytic paraganglioma' was first described by Kepes and Zacharias⁴ in 1971, because of features similar to ganglioneuroma and paraganglioma. Burke and Helwig⁵ reported 51 cases of gangliocytic paragangliomas in a review of the literature in 1989. We are reporting an interesting case of a gangliocytic paraganglioma who presented with an acute onset of left-upper quadrant abdominal pain that was treated with transduodenal excision and sphincteroplasty, with a subsequent excellent outcome.

CASE PRESENTATION

A 50-year-old man presented with an acute onset of left-upper quadrant abdominal pain with radiation to the back. He reported no nausea, vomiting or abdominal distension. He had no family history of gastrointestinal cancer or familial polyposis syndrome. He had a negative screening colonoscopy

Figure 1 CT revealed an intraluminal lesion in the second part of the duodenum.

1 year prior to presentation. Haemogram, liver function testing, amylase and lipase were all within normal limits. Abdominal CT revealed a 2 cm (cm) polypoid intraluminal mass at the junction of the first and second part of the duodenum (figure 1), with a normal pancreatic and hepatic parenchyma. The index upper endoscopy revealed a periampullary lesion that was partially occluding one-third of the lumen (figure 2). The initial biopsy was associated with profuse, self-limited bleeding, but was non-diagnostic with ulcerated smooth muscle on the background of acute chronic inflammation. MRI revealed a 2.4×1.9 cm periampullary mass along the medial second portion of the duodenum without an extra luminal component (figure 3). The pancreatic duct and common bile duct appeared normal in calibre and separate from the duodenal mass. Endoscopic ultrasound revealed a 3.0×1.6 cm, well circumscribed, hypoechoic duodenal lesion limited to the mucosa and parts of submucosa without regional adenopathy (figure 4). The common bile duct was normal and free of the lesion, while the pancreatic duct was poorly defined in the head of the pancreas. The pancreatic parenchyma was normal. Fine-needle aspiration (FNA) with a 25-gauge Wilson-Cook echo-tip needle was performed, followed by routine biopsies. The cytology revealed benign strips of glandular epithelium and ulcerative debris, while the biopsies revealed duodenal mucosa with endocrine

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Figure 2 Periampullary mass with the biliary and pancreatic orifice poorly visualised.

and epithelial cells, ranging in trabecular and nests with cells that were round, with clear to amphophilic cytoplasm, and round bland nuclei without mitosis. There were also spindle cells with abundant eosinophilic cytoplasm and elongated nuclei, which were positive for S100HU and synaptophysin (SYN B5), but negative for cytokeratin, all consistent with a gangliocytic paraganglioma.

DIFFERENTIAL DIAGNOSIS

1. Ampullary adenoma
2. Ampullary adenocarcinoma
3. Well-differentiated neuroendocrine carcinoma
4. Ganglioneuroma
5. Paraganglioma
6. Spindle cell malignant neoplasm (nerve sheath, smooth muscle and gastrointestinal stromal tumours)

Figure 3 MRI revealed an intraluminal periduodenal lesion. The pancreatic duct and common bile ducts were normal.

Figure 4 Endoscopic ultrasound revealed a periampullary lesion with mucosal and submucosal involvement (T1sm, No, Mo).

TREATMENT

The patient underwent exploratory laparotomy with wide transduodenal excision, with sphincteroplasty and cholecystectomy. Intraoperatively a duodenal mass was seen encroaching, but not involving, the common bile duct, while the pancreatic duct had a variant anatomy that opened separately and inferior in the ampullary region. The final pathology revealed a $3 \times 2.5 \times 1.6$ cm mass that was focally abutting the duodenum with free margin of resection. Immunohistochemistry were positive for SYN B5 and S100HU consistent with a gangliocytic paraganglioma.

OUTCOME AND FOLLOW-UP

The patient is followed with surveillance CT of the abdomen and pelvis, performed every 6 months, and upper endoscopy

Figure 5 Post wide surgical excision of ampullary lesion; patent biliary orifice.

every 6 months, have revealed no evidence of endoscopic or histological recurrence at now 3-year follow-up (figure 5).

DISCUSSION

The WHO classifies gangliocytic paraganglioma as a neuroendocrine neoplasm of the gastrointestinal tract.⁶ There is some controversy regarding the development of gangliocytic paragangliomas. The first theory suggests it originates from neuroectoderm/neurocrest cells.² The second theory suggests it originates from endodermal pluripotent progenitor cells.⁷ There are still some authorities who believe these tumours are hamartomas originating from the neuroectoderm and endoderm during embryonic development.⁸ The clinical presentation is variable and depends on the site of origin. The most common site is the periampullary region, but these tumours have been reported (rarely) in the appendix, jejunum, oesophagus, pylorus of the stomach, and even in the lungs.^{9–10} In most of the cases, the patients were asymptomatic. A review of 51 cases by Burke and Helwig⁵ reported abdominal pain, upper gastrointestinal bleeding with subsequent anaemia, and symptoms of obstructive jaundice as the most common presenting symptoms. Our patient presented with acute onset of abdominal pain, consistent with the Burke and Helwig⁵ review. The average age of presentation is 54 years with a male predominance.¹ Our patient was 50 years old, which is consistent with the literature. Gastrointestinal bleeding is secondary to mucosal and submucosal invasion.¹¹ In case of a submucosal location, endoscopic biopsy may not be diagnostic due to tumour location. In our case, the initial endoscopic biopsy was non-diagnostic, making preoperative diagnosis more challenging with simple endoscopy.¹² Grossly gangliocytic paragangliomas are usually polypoid (as in our case), sessile or pedunculated. The size varies from 0.5 to 10 cm in diameter.¹² The presence of three elements, epithelial, ganglionic cells and spindle cells, are mandatory for the diagnosis of gangliocytic paragangliomas.^{2–9} The presence of keratin in 50–60% epithelial cells is suggestive of a neuroendocrine origin and also differentiates them from paragangliomas.⁷ Immunohistochemistry studies reveal that spindle cells are positive for S100P, while ganglion cells are positive for synaptophysin and neuron-specific enolase.^{6–9} Our patient was positive for S100 and synaptophysin, but negative for cytokeratin, consistent with a gangliocytic paraganglioma. The size, location and benign versus malignant nature usually dictate the treatment approach.^{5–12} Endoscopic image-guided resection is the first treatment option, which includes simple resection or submucosal injection of saline solution followed by endoscopic mucosal resection.^{9–13} In our patient, the benign nature of the tumour and the unique anatomy of a separate opening of the pancreatic duct and common bile duct lead to a transduodenal excision with sphincteroplasty, thereby avoiding extensive surgery such as a Whipple procedure. A partial pancreaticoduodenectomy is the best option for

periampullary malignant gangliocytic paragangliomas and obstructive jaundice.

Learning points

- ▶ Endoscopic ultrasound fine-needle aspiration (EUS-FNA) leads to preoperative diagnosis of gangliocytic paraganglioma benign versus malignant so proper treatment strategy can be tailored.
- ▶ EUS-FNA-guided biopsy can avoid open surgery as benign lesions can be treated by simple resection and electronic medical record.
- ▶ Partial pancreaticoduodenectomy is the best option for periampullary malignant gangliocytic paragangliomas with obstructive jaundice.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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