Unusual presentation of more common disease/injury

CASE REPORT

Gastrointestinal stromal tumour masquerading as a cyst in the lesser sac

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SUMMARY

Gastrointestinal stromal tumours (GISTs) are solid tumours of the gastrointestinal tract, mostly found in the stomach and intestine. They rarely present as cystic lesions. A 74-year-old woman referred to the hepatopancreatobiliary unit, with 3 months history of upper abdominal discomfort. Abdominal ultrasound scan showed a large cystic lesion in the epigastric region suggestive of a pancreatic pseudocyst. The CT-scan showed a 6.6×6×6.3 cm size cyst related to the pancreas and extending to the hepatico-gastric omentum. Endoscopic ultrasound (EUS) scan was suggestive of a pancreatic pseudocyst. Aspirated Cyst fluid via EUS showed benign cytology with normal amylase, lipase and tumour markers (CEA, CA-19.9 and CA-125). She was referred as a case of pancreatic pseudocyst. After surgical excision, the histopathology confirmed the presence GIST in the wall of the cystic lesion. The possibility of GIST should be kept in mind in the presence of unusual features of a cyst on abdominal imaging.

BACKGROUND

We report this case because of the diagnostic dilemma we faced during the initial investigations, both radiological and laboratory. The histopathological examination of the specimen resulted in an unexpected diagnosis. Accordingly, further surgical intervention was needed.

CASE PRESENTATION

A 74-year-old patient presented to the surgical outpatient department of the hepatopancreatobiliary unit, referred from a district general hospital, with 3 months history of upper abdominal pain and discomfort, dull aching in nature and radiating to the back and left upper quadrant. It was associated with nausea but no vomiting. There was no change of bowel habit, no loss of weight or appetite. She was also diabetic (type II) on oral hypoglycaemic, hyper-tensive with ischaemic heart disease. Abdominal examination showed non-tender non-pulsatile epigastric region fullness with ill-defined edges.

INVESTIGATIONS

An ultrasound scan, performed 2 months prior to the time of presentation, showed a large cystic lesion in the epigastric region with turbid fluid content, suggesting that it might be a pancreatic pseudocyst. However, the patient did not show any history suggestive pancreatitis. Her laboratory investigations showed normal liver and kidney functions with mild anaemia (haemoglobin 11 gm/dL). An abdominal CT scan showed a cyst related to the pancreas measuring 6.6×6×6.3 cm (figure 1A, B). In addition there were other incidental findings such as, bilateral small adrenal adenomata, and a small liver cyst in the right lobe which had no blushing or wall enhancement in the delayed CT images and was deemed a simple liver cyst by the radiologist. Tumour markers including CEA, AFP, CA-19.9 and CA-125 were all within the normal range. Echinococcus granulosus serology was negative. The patient underwent an endoscopic ultrasound assessment with guided fine-needle aspiration cytology. The impression was that of a pancreatic cyst measuring 6×8 cm. The cystic of the cystic fluid was benign and both amylase and lipase levels in the fluid were within the normal range measuring 23 and 24 IU/L, respectively. CEA and CA-19.9 levels in the fluid were within the normal range (1.0 μg/L and 3.84 IU/mL, respectively). The microscopic examination of the fluid revealed no hydatid hooklets. Culture of the cyst fluid showed no growth of any microorganisms.

DIFFERENTIAL DIAGNOSIS

▸ Pancreatic cyst.
▸ Simple liver cyst.
▸ Hydatid cyst.
▸ Duplication of bowel (stomach).

TREATMENT

A multidisciplinary meeting including general surgeons, endoscopists and radiologists was held before the first operation. The diagnosis of a pseudopancreatic cyst was highly doubtful according to the surgeons. On reviewing the CT images with the radiologists, there was no evidence of a connection between the cyst and the pancreas which looked healthy. There was a high suspicion of exophytic liver cyst or bowel duplicate.

The patient underwent diagnostic laparoscopy which showed a thin-walled cystic lesion, freely mobile within the lesser (hepatogastric) omentum with no attachment to the liver or pancreas, but part of its wall (about one-eighth) was attached to the lesser curvature of the stomach. Aspirate from this cyst revealed a brownish thin fluid which was sent for cytology and tumour markers (both were negative) test. The cyst was evacuated and the thin wall was excised leaving the part which is attached to stomach. A drain was inserted and the procedure was terminated awaiting the histopathological examination of the excised part of the wall.

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OUTCOME AND FOLLOW-UP

The pathology report revealed an unexpected diagnosis of gastrointestinal stromal tumour (GIST). The case was discussed in our biweekly multidisciplinary oncology meeting in the surgical department in the presence of histopathologists and oncologists. It was agreed in the meeting that the residual tumour should be excised and to start imatinib therapy after the second operation. The patient was recalled and she underwent laparotomy through a bilateral subcostal incision. The remaining part of the tumour together with the adherent parts of lesser omentum and lesser curvature of the stomach were excised within safety margins. In addition, pyloroplasty was carried out in the gastrohepatic (lesser) omentum. In the coronal cuts, the cyst (6.6×6×6.3 cm) between the left lobe of the liver and the stomach, always considered of GIST. After excision with wedge resection of the greater curvature of the stomach, where the stalk was attached, histopathological examination revealed GIST. They suggested that in cases of unusual exophytic pendulated tumour of the stomach, always consider of GIST.

Cystic GIST lesions are rare and invariably diagnosed late after excision. In 2012 De Vogelaere et al reported a large (12 cm) exophytic cystic tumour with no preoperative diagnosis of its origin. After excision with wedge resection of the greater curvature of the stomach, where the stalk was attached, histopathological examination revealed GIST. The authors suggested that in cases of unusual exophytic pendulated tumour of the stomach, always consider of GIST.

In 2014 Hansen et al reported a 74-year-old female patient who underwent a Roux-en-Y cyst-jejunostomy for pancreatic pseudocyst. Few weeks postoperatively, she developed several melena episodes with negative upper GI endoscopy. She was surgically reappraised. The main diagnostic concern was a pancreatic cystic neoplasm. A 12×8.0×5.0 cm size retrogastric lesion was resected and pathology report indicated an unsuspected GIST, which was previously misdiagnosed as a pancreatic pseudocyst.

Cystic GIST tumours may be observed as a primary cystic GIST, in which the main structure comprises a cystic tissue with a pseudocapsule, or a rapidly growing malignant GIST with central cystic degeneration due to insufficient blood supply resulting in necrosis and liquefaction. When the tumour metastasises to the liver and pancreas, the metastatic lesion is always cystic in nature, often confused with liver cysts and pancreatic cysts. In addition, with imatinib treatment malignant GISTs may show cystic degeneration.5

Cystic GIST lesions became a distinct entity after the discovery of KIT (CD11) in 1998. GISTs originate from the intestinal pace-maker cells of Cajal, therefore are usually confined to the submucosa and muscularis propria.1 It affects both sexes equally and 80% occur in persons over 50 years. They arise as a result of oncogenic mutation in the KIT tyrosine kinase. Most (75–80%) GISTs have KIT mutations, typically affecting the juxtamembrane domain encoded by exon-11. These tumours account for 1% of all intestinal neoplasms, the age-adjusted incidence in Europe and the USA is 7 cases per million.2 Independent adverse prognostic factors are large tumours, high mitotic count, non-gastric location, rupture and male gender.2 4 GISTs can arise anywhere in the GI tract, but most commonly in the stomach and small intestine.2 About 60% of patients are cured by surgery. After complete excision, it may recur within 3 years in approximately 50% of patients. Imatinib is recommended for patients with substantial risk of recurrence.4

This patient was referred as a case of pseudocyst of the pancreas although the patient never had any previous attack of pancreatitis. On reviewing the images, the pancreas was normal with no radiological evidence of pancreatic abnormality denoting previous pancreatitis and there was no connection with the cyst. To our surprise the first histopathology reported GIST tumour. GISTs are usually solid tumours, however, they may rarely present as cystic lesions.

In the same year, Zhu et al reported an exophytic retrogastric large cystic lesion which they reported to have a similar diagnostic dilemma, even after a CT scan and MRI. It was thought to be a pancreas-related cyst. After block excision of the mass with spleen, greater omentum and the attached wall of the stomach the histopathological examination revealed GIST. They suggested that the use of ultrasound-guided endoscopy might have provided further diagnostic evidence.5 However, we can say from our experience that the use of endoscopic ultrasound did not help that much in a relatively similar case. It helped only in the exclusion of other pathologies.

Recently in 2015, Kumar et al reported a 55 year old woman who presented with a painful lump in the epigastrium. A CT...
scan revealed a large exophytic cystic lesion from the duodenum measuring 15×10×8 cm arising from the lateral wall of the second part. Preoperatively, it was thought to be a duodenal diverticulum. After resection, and pathological examination they were surprised to find that it was a duodenal GIST tumour.

Our case posed a diagnostic difficulty as it was thought to be either a pancreatic pseudocyst or a liver exophytic cyst. Intraoperatively, it had no attachment to any of the two organs. The thick posterior wall however was suspicious. We considered the possibility of organ duplicate, which has been occasionally reported. To our surprise, it turned out to be GIST. The literature search of the last 5 years revealed the aforementioned cases which had the same diagnostic dilemma. We conclude that GIST should be considered one of the differential diagnoses when any bowel-related cystic lesion is encountered.

**Contributors** RA provided the preoperative workup results at al Jahra Hospital. AMH wrote the manuscript draft. SA and EHA reviewed it. The histopathology and the immunostain microphotographs were provided by IF. The final manuscript was reviewed and approved by all authors.

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