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A rare presentation of small diaphragmatic epidermoid cyst with extremely elevated serum CA19-9 level

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ABSTRACT

Epidermoid cysts are rare lesions that can occur anywhere in the body. They are associated with elevated serum levels of CA 19-9. The spleen represents the most common site of intra-abdominal localisation. Only two cases of diaphragmatic epidermoid cyst are reported in the literature. We present the case of a 61-year-old woman with a small suprasplenic subdiaphragmatic cyst discovered during the investigation of left flank pain. The establishment of an adequate diagnosis was challenging due to the difficulty in specifying the exact localisation of the cyst, the extremely elevated CA 19-9 level of 19,000 and the high uptake on 18-fluoro-2-deoxy-D-glucose positron emission tomography. The definitive diagnosis followed complete surgical excision. Intra-abdominal epidermoid cysts are usually discovered incidentally on imaging for another reason. The cyst is lined by squamous epithelium responsible for the secretion of CA 19-9. The elevation of serum CA 19-9 is due to small rupture or increased intraluminal pressure followed by diffusion to the bloodstream. Surgery with en-bloc resection represents the optimal treatment to avoid any risk of recurrence. The definitive diagnosis is established by demonstrating positive immunohistopathological staining of epithelial cell to CA 19.9.

KEYWORDS

Diaphragmatic epidermoid cyst – CA 19-9 level – FDG-PET scan – En-bloc resection – Immunohistochemistry

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Background

Epidermoid cysts are rare lesions that can grow in multiple regions in the body (palm, oral cavity, middle ear, neck, kidney, gastrointestinal tract, diaphragm and testis, head, brain).[1–3] Independent of their location, epidermoid cysts are associated with increased serum levels of the tumour biomarker carcinoembryonic antigen (CA) 19-9.[1–3] Diaphragmatic epidermoid cysts are extremely rare. To our knowledge, only two cases have been reported in the literature.[2,3] The exact localisation is difficult to define based on preoperative imaging and is usually mistaken for liver or splenic origin. We present a case of challenging diagnosis of left diaphragmatic epidermoid cyst with an extremely elevated CA 19-9 level.

Case history

A 61-year-old woman with a history of kidney stones presented with left upper quadrant abdominal pain, colicky in nature, moderate in intensity and associated with nausea. Her physical examination was positive for left flank tenderness. Urinalysis was negative and the blood test showed a haemoglobin of 14.2 g/dl and white blood cell count of 7500 cells/µl. A kidney ultrasound showed a subdiaphragmatic heterogeneous cystic lesion (Fig 1). Abdominal computed tomography showed a left subphrenic suprasplenic mass measuring 29 × mm× 21 mm (Fig 2). The exact origin of the mass could not be specified with certainty.

Laboratory examination showed normal amylase, lipase and liver function tests. Tumour biomarkers showed

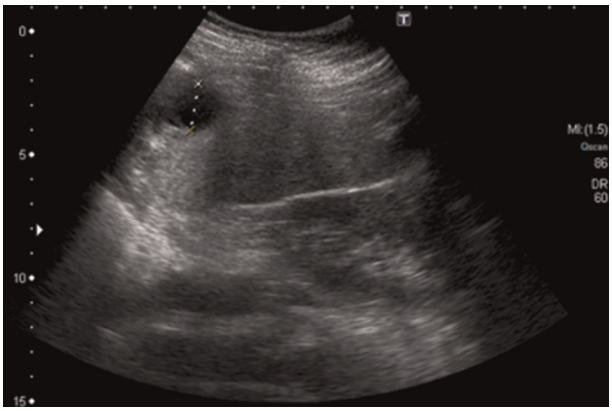

Figure 1 Abdominal ultrasound examination showing the well-defined cystic lesion, a gastric antral lesion.

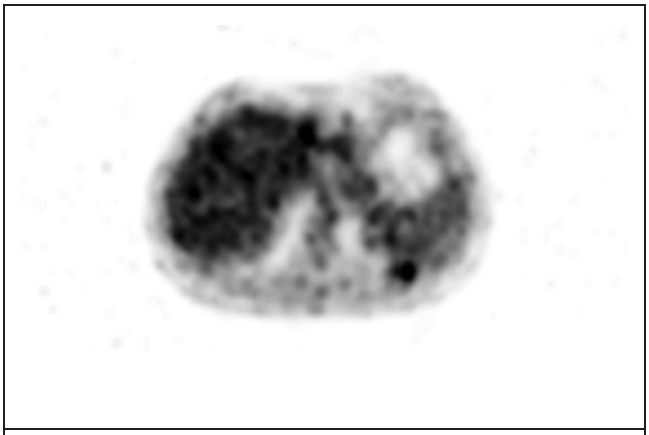

Figure 3 18-fluoro-2-deoxy-D-glucose (FDG) positron emission tomography demonstrating high FDG uptake in the cystic lesion (maximum standard uptake value 3.34).

normal alphafetoprotein, CA 125 and elevated CA 19.9: 12000 kU/L (normal < 37 kU/L). The CA 19-9 test was repeated after four days and showed an increased level to 19000 kU/L. 18-fluoro-2-deoxy-D-glucose positron emission tomography (FDG PET) demonstrated solitary heavy uptake in the wall of the lesion (maximum standard uptake value 3.34; Fig 3). As a result of the elevated CA 19-9 and the hypermetabolic uptake shown on FDG PET, the patient was consented for exploratory laparoscopy and cyst excision. The exploration showed no peritoneal carcinomatosis and no liver metastasis. The splenic angle was mobilised by incision of the left gastrocolic ligament and to the left line of Toldt. The spleen was mobilised medially by division of the splenophrenic and the splenocolic ligaments. Exploration of the left retrosplenic space showed the diaphragmatic lesion, which was completely independent of the spleen.

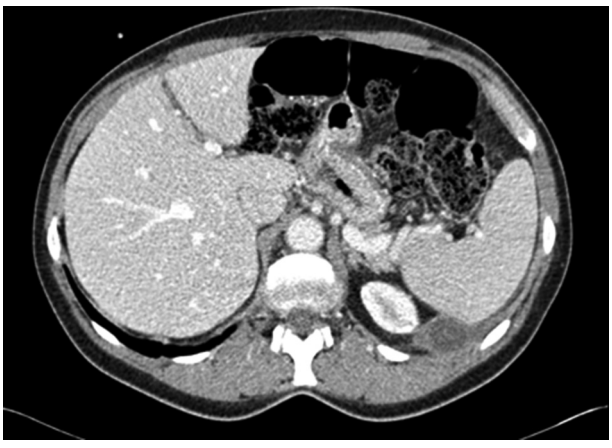

Figure 2 Abdominal computed tomography showing the 29 × 21 mm suprasplenic, subdiaphragmatic cystic lesion.

Owing to irregular margins and difficult exposure, the surgery was converted to laparotomy, with partial resection of the diaphragm and en-bloc resection of the lesion. The diaphragmatic defect was closed with interrupted vicryl 2 sutures on a tube after inflation of the lung. A frozen section suggested a benign ciliated foregut cyst. The patient had an uncomplicated postoperative recovery and she was discharged home at postoperative day 3. The definitive pathology showed a 2-cm epidermoid cyst. The epithelial lining was squamous in origin and stained positive for cytokeratin (CK) and CA 19.9 and negative for calretinin and CD34 with no signs of malignancy (Fig 4). CA 19-9 levels were taken three months postoperatively and had returned to normal (CA 19.9 30 kU/L. The patient remains recurrence free seven years post-surgery.

Discussion

Diaphragmatic lesions are rare entities. Their differential diagnosis comprises a variety of benign conditions such as cysts, lipomas and leiomyomas and malignant lesions, which are predominantly sarcomas and mesothelioma.[4] Diaphragmatic epidermoid cysts represent the least frequent subset of diaphragmatic cysts after mesothelial and bronchogenic cysts with, to our knowledge, only two cases of diaphragmatic epidermoid cysts reported in the literature.[2,3] As with abdominal epidermoid cysts, most patients are asymptomatic at presentation and the lesion is discovered incidentally. Patients might present with symptoms related to mass effect such as abdominal pain, nausea, vomiting and dyspnoea.

The exact localisation of the cyst is sometimes challenging and is defined based on preoperative imaging that specifies the size, density, anatomic localisation and relation to organs and to vessels.[4] In our case, similar to the two reported cases of diaphragmatic epidermoid cysts, preoperative CT was not sufficient to localise the cyst and its diaphragmatic origin was identified intraoperatively.[2,3]

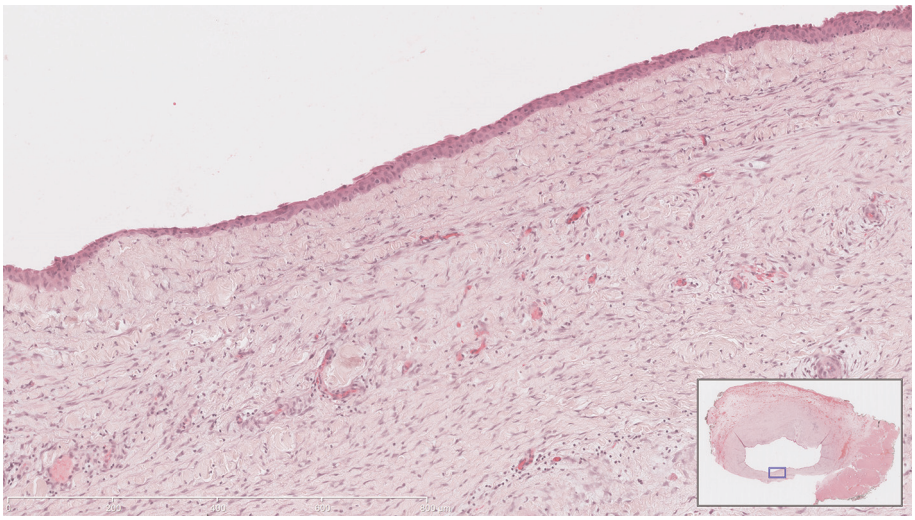

Figure 4 Histological image of the cyst lined by squamous epithelium (haematoxylin-eosin staining).

In our patient, initially after considering that the cyst might be of splenic origin, the serum tumour markers showed a raised CA 19-9 level and repeated laboratory tests confirmed this high value. The CA 19-9 level is usually a tumour marker for pancreatic among other types of gastrointestinal adenocarcinomas. It might be elevated in cases of non-malignant biliary conditions such as choledocholithiasis and cholangitis and in cases of benign cystic lesions such as mediastinal, bronchogenic and splenic cysts.[5] The serum CA 19-9 level was elevated in one of the two reported cases[2] but not in the other.[3] The level of CA 19-9 decreased to normal after the successful removal of the cyst. FDG PET was performed to evaluate occult intrapancreatic lesions and showed (similar to previous publications) a high uptake in the cystic lesion with no other focus of hypermetabolism.[2] It remained unclear why this lesion showed hypermetabolic uptake on FDG PET, since no signs of inflammation were present at histopathological examination. Abdominal epidermoid cysts are best treated by laparoscopic surgery with en-bloc excision of the cyst and partial or complete resection of the containing organ.[2–4] Surgery is indicated in case of symptoms or enlargement of size greater than 5 cm because of the risk of rupture. Less invasive operations such as drainage with marsupialisation are associated with a high risk of peritoneal fluid dissemination and risk of recurrence . The definitive diagnosis is based on the pathological examination of the resected specimen. Lining epithelium is of squamous origin and stains positive for CA 19-9 and cytokeratins on immunohistochemical examination and is negative for CD34 and calretinin.[1–5] High concentrations of CA 19-9 have been found in the fluid drained from epidermoid cysts.[2] Unfortunately the fluid was not examined in our case.

Conclusion

Epidermoid cysts are rare benign lesions associated with elevated CA 19-9 levels. Surgery is indicated in case of symptomatic presentation or increase in size and if there is a suspicion of malignancy. The definitive diagnosis is based on histopathological examination of the resected specimen and the causal relation with the elevated CA 19-9 level is confirmed after its postoperative normalisation.

References

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