Adult gastric duplication cyst: diagnosis by endoscopic ultrasound-guided fine-needle aspiration (EUS-FNA)

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ABSTRACT

Duplication cysts are rare gastrointestinal congenital abnormalities. They can occur anywhere throughout the gastrointestinal tract, and gastric duplication cysts are most uncommon, representing only 4-8% of all gastrointestinal duplication cysts. Nowadays several theories try to explain the pathogenic mechanisms involved. These cysts are usually diagnosed during early childhood, and very rarely are detected in adults, mostly incidentally due to a lack of symptoms. Close to 50% of gastric cysts are associated with other abnormalities. It is extremely important that a meticulous differential diagnosis is performed regarding other diseases, mainly malignancies with a cystic component. Although extremely uncommon, a malignant transformation of these lesions has been reported, which highlights the importance of a correct diagnosis. Herein we report the case of a duplication cyst in an adult, which was detected by endoscopic ultrasound-guided fine-needle aspiration, recently proposed as the most accurate technique for the identification of these lesions.

Key words: Gastric duplication cyst. Endoscopic ultrasound (EUS). EUS guided fine-needle aspiration (EUS-FNA).

RESUMEN

Los quistes de duplicación son anomalías congénitas infrecuentes del tracto gastrointestinal que pueden localizarse a cualquier nivel desde la boca hasta el ano. Los quistes de duplicación gástricos son los más excepcionales y constituyen tan sólo el 4-8% de todos ellos. Su patogénesis es controvertida y hoy en día aún existen múltiples teorías que tratan de explicar su mecanismo etiopatogénico. En la mayoría de los casos el diagnóstico se realiza en la primera infancia y tan sólo en una pequeña proporción se identifican en la edad adulta como un hallazgo incidental, ya que estas entidades suelen ser asintomáticas fuera de la infancia. Casi en la mitad de los casos se asocia a otras malformaciones congénitas y es de vital importancia realizar un minucioso diagnóstico diferencial con otras entidades, en especial neoplasias con componente quístico. Aunque es extremadamente inusual, se ha publicado la degeneración neoplásica de estas lesiones, lo que hace más importante su preciso diagnóstico y estudio citohistológico. En este artículo se presenta el caso de un quiste de duplicación gástrico diagnosticado en la edad adulta, mediante ultrasonografía endoscópica asociado a punción-aspirativa, técnica que ha sido recientemente propuesta como la más eficaz para el diagnóstico.

Palabras clave: Quiste duplicación gástrico. Ultrasonografía endoscópica (USE). Punción aspiración guiada por ultrasonografía endoscópica (USE-PAAF).

INTRODUCTION

Duplication cysts are extremely uncommon lesions. They are congenital abnormalities and their prevalence is very low. They may be potentially located anywhere along the gastrointestinal tract. Gastric duplication cysts, together with appendicular ones, are most uncommon.

Diagnosis is made in more than half of cases during early childhood, because duplication cysts tend to be symptomatic at this age. Conversely, these cysts are usually asymptomatic during adulthood, and their diagnosis is incidental. In almost half of cases they are associated with other malformations, mainly located in the esophagus and vertebrae. Duplication cysts are not exempt from developing complications such as bleeding, fistulization, and even malignant degeneration. Imaging techniques such as CT and MRI do not accurately diagnose these lesions, and perform rather poorly in terms of lesion characterization. On the other hand, recent reports suggest that endoscopic ultrasonography (EUS) may play a major role in the diagnosis of this disease, showing higher accuracy rates versus traditional imaging techniques.

There is no worldwide-accepted therapeutic algorithm for duplication cysts. Surgery is recommended for symptomatic cases or when a complication arises. However, to date, there is no consensus on the management of asymptomatic cases. This is seminal especially if we consider the risk of malignant transformation.

Duplication cysts were first described by Wendel in 1911, and few cases have been reported since then. Therefore, we report the case of a gastric duplication cyst in an adult patient, as well as a review of the literature.

CASE REPORT

A 39-year-old male was referred to our Echoendoscopic Ultrasound Unit for evaluation of a submucosal lesion located in the gastric side of the esophagogastric junction. The patient was visited in the medical service of Otorhinolaryngology for throat discomfort. The patient had an upper endoscopy that showed a 15-mm elevated lesion with preservation of the mucosa, located 36 cm away from the dental arcade. The lesion was biopsied, but the pathological analysis of the specimen was inconclusive. Due to the lack of a precise diagnosis, the patient was referred to our center for further evaluation with EUS.

Endosonography was performed under vital sign monitoring, conscious sedation with midazolam and pethidine, and on an outpatient basis. A linear Pentax echoendoscope EG3630UX was used coupled to a Hitachi 8500 ultrasound probe. At 36 cm from the dental arcade we identified a small cystic anechoic lesion measuring 14 x 6 mm (Figs. 1 and 2), apparently dependent on the mucosal muscle. Neither abdominal nor mediastinal enlarged lymph nodes were identified. EUS ensured the integrity of the vascular axis, and Doppler sonography was used to locate the optimal access for EUS-guided puncturing with an Echotip Wilson-Cook 19 G needle. The needle sheath was removed a few centimeters from the working channel. Afterwards we proceeded to puncture the lesion with the needle located inside the sheath. Once verified our position inside the lesion, the stylet was withdrawn (it is
used to prevent potential sample contamination) and a puncture was made with back and forth movements in order to maximize the amount of tissue obtained. We also used syringe aspiration to increase the cellularity of the sample. Two passes on the lesion were needed to confirm the adequacy of the material by a pathologist present during the exploration.

The Diff-Quick (Merck) stain was used to evaluate the specimen in situ, this being later supplemented with a cytological study using Papanicolaou’s stain (Merck). The study showed abundant histiocytes and cylindrical epithelium suggestive of a benign cyst consistent with a gastric duplication cyst (Figs. 3 and 4).

**DISCUSSION**

Duplication cysts are cystic formations adjacent to a portion of the gastrointestinal tract with which they share a wall. They are lined by a mucosa similar to that in the adjacent structures. They are low-prevalence congenital abnormalities that can be located at any level in the gastrointestinal tract. In descending order of frequency, they may be located in the ileum, esophagus, jejunum, colon, stomach, and appendix (1,2). Therefore, a gastric location is extremely unusual and represents only between 2 and 8% of all these entities (1-3).

These structures are the result of an altered embryonic development. Several etiopathogenic theories have been proposed to explain their formation: a) persistence of a vacuole formed in the solid phase of bowel embryogenesis or persistence of an embryonic diverticulum; b) failed fusion and recanalization of the longitudinal folds, which would allow an epithelial bridge; and c) formation of a traction diverticulum as a result of a failure in the normal development of the notochord and endoderm. Despite all these theories, the etiopathogenic origin of duplication cysts remains unclear (1,2,4). Duplication gastric cysts originate dorsal to the primitive intestine, so most of them are located in the greater curvature (1,2), and only 5.5% of them are in the smaller curvature (1).

The prevalence of duplication cysts is twice as much in women, and they show no familial aggregation (1,3,5). They are usually diagnosed at an early age (more than 60% of cases in the first year of life), so a diagnosis in an adult is extremely rare (4,6). Clinical symptoms depend mostly on patient age, and lesion location and size. In the adult they are often asymptomatic, and, as happened in our case, the diagnosis is made incidentally during a study indicated for a non-related condition.

Recurrent abdominal pain, a palpable mass, vomiting, decreased weight or failure to thrive characterize the symptomatic form of this disease. Other possible events are related to the development of complications such as fever (due to cyst overinfection), gastrointestinal bleeding, fistulization, twisting of its pedicle, and intestinal occlusion (2,3,5). A case of a duplication cyst in the pylorus of a newborn simulating benign hypertrophy of the pylorus has been reported (3). In half of cases there are associated malformations, most frequent of these being esophageal duplication, followed by vertebral abnormalities (1,2,5).

In 1959 Rowling described a number of typical characteristics of duplication cysts, such as proximity to the digestive tract, a common blood supply, a layer of smooth muscle shared with the gastrointestinal wall, and gastrointestinal epithelium lining (7). However, some reports include heterotopic mucosa (respiratory) and pancreatic epithelium (8,3,9). The presence of pancreatic or gastric mucosa is associated with complications such as...
peptic ulcer or acute pancreatitis (2,3). Although infrequent, malignant transformation has been reported, usually in the form of adenocarcinoma (10) and squamous cell carcinoma. Also, and even more infrequent, duplication cysts can degenerate into neuroendocrine carcinomas (11,12).

Differential diagnosis must include other cystic neoplasms or tumors with cystic degeneration (e.g. intraductal papillary mucinous tumor, mucinous cystadenoma, lymphangioma), and other benign lesions – e.g. simple cyst, lymphoepithelial cyst, pancreatic pseudocyst, inclusion cysts, or parasitic cysts (4,6). In summary, in order to rule out malignant cyst transformation, a cytological and histological examination of the lesion is mandatory.

Abdominal CT and MRI can identify duplication cysts, but misdiagnosis of solid masses can reach up to 70% depending on the series (13). The main source of misdiagnosis comes from the variable content and the presence of highly concentrated proteins in some of these cysts. USE is crucial for the diagnosis and characterization of submucosal lesions (14). It allows to define with high precision the relation between the cyst wall and the adjacent gastrointestinal structures. It also helps differentiate between solid and cystic lesions. To our knowledge, there are nine studies published that specifically assess the role of EUS in the diagnosis of duplication cysts (13,15-22), including three cases with gastric location (15,20,22). Most reports show that EUS is a safe and accurate technique for the diagnosis of this type of lesion. USE also allows to obtain tissue for cytopathological examination, which is essential for the differential diagnosis with other entities, as well as to exclude malignant transformation of the cyst. Although studies are fairly heterogeneous, it seems that cytology is the key technique for the diagnosis of these lesions (13). Robust evidence supports the fact that EUS has higher sensitivity rates than conventional endoscopy for the characterization of submucosal lesions (14). Therefore, EUS, and above all, EUS-guided fine-needle aspiration has been proposed as a first-line technique for the diagnosis and characterization of duplication cysts. Most cases published show a cyst size superior to ours, from 2 cm (3) up to 15 cm (11). The fact that there has been punctured a lesion of cystic characteristics of such small size could be subject of discussion.

There is no worldwide-accepted diagnostic and therapeutic algorithm for duplication cysts. Surgical treatment is recommended for symptomatic cases, although the optimal surgical procedure remains controversial. Some authors also recommend the surgical removal of the lesion in asymptomatic cases, arguing that this would prevent future complications and the risk of malignant degeneration (2,6,11,23). Conversely, other authors favor conservative treatment, based on the fact that these lesions are incidental in adults, and that malignant transformation is anecdotal (13). Interestingly, a case of spontaneous resolution of a duplication cyst in the mediastinum has been reported (24). Zambudio et al. proposed several criteria for the surgical excision of mediastinal cysts: presence of symptoms, infection, compression of the trachea, progressive growth, diagnosis in childhood, and atypical features or location (23). Several authors argue that some of these criteria could be extrapolated to the management of gastric duplication cysts, although specific studies should be conducted to evaluate this and other therapeutic options.

REFERENCES