Congenital agastria as an isolated malformation

Agastria congénita como una malformación aislada

Congenital agastria is an extremely rare condition\(^1\) that is frequently associated with other gastrointestinal and extra-gastrointestinal tract malformations.\(^2\)-\(^4\) We present herein the case of a female patient with congenital agastria, with no other associated malformation.

A female infant of 2 months of age was the product of a second pregnancy of 34-week gestation. She was delivered by cesarean section, weighed 2,000 g (-3.1 SD, p0) and measured 43 cm in height (-3.3 SD, p0), corrected for gestational age. The infant had a history of necrotizing enterocolitis at one month of life and was referred to the Gastroenterology and Nutrition Service of the Instituto Nacional de Pediatría for vomiting and oral diet intolerance. She was managed with prokinetics and exclusion of cow-milk protein, with no improvement. Upon admission, physical examination showed no alterations, with weight of 2.490kg (-5.37 SD, p0) and height of 44 cm (-6.42 SD, p0). Contrast-enhanced esophagogastroduodenoscopy revealed dilation of the body of the esophagus and a tubular gastric chamber (fig. 1a). Abdominal ultrasound showed esophageal dilatation (transverse diameter: 15 mm) and reduced gastric capacity (10ml). Congenital microagastria was suspected. Dilation of the esophageal body was observed in the esophagogastroduodenoscopy, with an absence of epithelial change in the esophagogastric junction and the folds of the gastric corpus. An atrophic and pale mucosal remnant was also observed that was first interpreted as the antral region (fig. 1b). Biopsies of the site were taken that reported superficial esophageal epithelium with hydropic degeneration, supporting the diagnosis of congenital agastria. Echocardiogram, transfonnetellar, abdominal, and kidney ultrasound studies, kidney function tests, and x-rays of the spine and extremities were performed to rule out the coexistence of other congenital malformations. A diet based on formula with initial continuous infusion was begun and was adequately tolerated. Once the patient achieved a certain weight recovery, she underwent the creation of a Hunt-Lawrence pouch at 3 months of age. During the intermediate postoperative period, the patient presented with abdominal sepsis. K. pneumoniae BLEE and E. faecalis were isolated and the infant was given broad-spectrum antibiotic therapy. At present, she is 6 months old and is adequately tolerating oral diet and gaining weight in accordance with her corrected gestational age: weight: 5.83 (-0.78 SD, p22), height: 62.2 (0.05 SD, p52).

Congenital agastria is the result of an alteration in the embryogenesis of the stomach. The process begins at the 5th week of fetal life, with the appearance of the gastric primordium (located at the distal part of the anterior intestine), which will later give rise to the stomach. Depending on the time at which that process is interrupted, either complete absence of the stomach or the formation of a small, tubular gastric remnant with minimal functional capacity (microgastria) will be produced.\(^5\) The clinical data that those patients commonly present with are postprandial vomiting, gastroesophageal reflux, aspiration pneumonia, and malnutrition. Symptoms vary depending on the phase at which the development of the stomach was detained.\(^2\)

As previously mentioned, those disorders often are accompanied by other gastrointestinal (esophageal atresia, intestinal malrotation, asplenia, imperforate anus), cardiac, renal, and skeletal abnormalities. Therefore, the initial approach must include the identification of those anomalies.\(^2\)-\(^4\)

Diagnosis is suspected through esophagogastroduodenoscopy that commonly reveals a tubular gastric remnant in the midsagittal position, as well as esophageal dilation. However, it is important that biopsies be taken at esophagogastroduodenoscopy, given that the differential diagnosis between agastria and microgastria cannot be made through
radiologic or endoscopic images, as was seen with our patient.

Initial treatment is usually medical and consists of ensuring an adequate fluid and electrolyte status and aiding the nutritional recovery of those patients. Nutritional strategies employed for that purpose have been noninvasive (nutrition through continuous infusion utilizing a nasogastric tube) and even surgical (placement of gastrostomy or jejunostomy catheters).2,5,6

The creation of a food reservoir has been shown to be effective for enabling adequate weight gain and improving quality of life in those patients. It can be achieved through the creation of a Hunt-Lawrence pouch, in which an efferent jejunal segment is anastomosed in a laterolateral manner to an afferent jejunal segment, forming a proximal Roux-en-Y pouch.7-9 That technique enables an increase in gastric capacity, reducing the frequency of intakes, improving nutritional ingestion, facilitating bowel transit, and preventing alkaline reflux esophagitis.7 Until 2010, only 13 cases managed through that surgical technique had been reported in the medical literature.1 In cases of isolated congenital agastria, some authors recommend early performance of said procedure, given that it is highly unlikely that the stomach will spontaneously grow.9 Postprandial symptoms during the intermediate postoperative period, such as dumping syndrome, epigastric pain, steatorrhea, fat malabsorption, and bacterial overgrowth, have been reported.4,10 We observed none of those symptoms in our patient. In addition, pernicious anemia prophylaxis with monthly vitamin B12 administration is recommended, due to the lack of intrinsic factor production.

To the best of our knowledge, ours is the first well-documented case of congenital agastria with no other congenital malformations.

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Conflict of interest

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References


C.A. Cuadros-Mendoza *, M.C. Martinez-Soto, F.E. Zarate-Mondragón, R. Cervantes-Bustamante, J.A. Ramirez-Mayans

Servicio de Gastroenterología y Nutrición, instituto Nacional de Pediatría, Mexico City, Mexico
Anemia as initial presentation of lung cancer with metastasis to the small bowel

Anemia como presentación inicial del cáncer de pulmón con afectación intestinal metastásica

Metastatic gastrointestinal lesions from the lung can appear at any part of the gastrointestinal tract and usually present at advanced stages of the disease, conferring poor prognosis. Duodenal appearance is exceptional and jejunal involvement implies a greater risk for perforation.1,3

We present herein two cases of patients with anemia due to metastasis to the small bowel as the initial presentation of lung cancer.

The first case corresponds to an 86-year-old man, ex-smoker, admitted to the hospital for constitutional symptoms and asymptomatic anemia. Complementary imaging studies (ultrasound and abdominal computed tomography [CT]) identified nonspecific duodenal involvement. At endoscopy, the duodenum showed signs of neoplastic infiltration. The histologic study revealed a poorly differentiated non-small cell carcinoma of pulmonary origin. Chest CT identified a 15 x 19 x 20 mm spiculated nodule in the left upper lobe suggestive of a primary tumor of the lung (positive CAM 5.2 and TTF1) (fig. 1 A). Given the age of the patient, his comorbidity, and quality of life based on a previous Eastern Cooperative Oncology Group (ECOG) grade of 3-4, palliative management was decided upon and the patient died 4 months later.

The second case was a 68-year-old man, ex-smoker, who presented with severe iron deficiency anemia. Capsule endoscopy revealed angiodysplastic lesions in the jejunum with active bleeding, along with possible jejunal stricture (fig. 1B and C). CT confirmed jejunal neoplasia with lymph node involvement and liver metastases. Further study included a chest CT scan and positron emission tomography (PET) scan, confirming the presence of disseminated disease with a primary tumor of the lung (stage IV). Immunohistochemical analysis of the biopsies identified the adenocarcinoma subtype (positive CAM 5.2; negative CDX-2 and keratin 20; positive TTF1 and keratin 7) (fig. 1D). Given the patient’s good general condition, chemotherapy was begun (carboplatin [CBDCA] + Alimta + atezolizumab) with good response (survival at 5 months from diagnosis).

Lung cancer is the first cause of death by cancer in the Spanish environment and 50% of the cases develop metastasis. The most frequent subtype is non-small cell carcinoma (85%).1,4-8

The gastrointestinal tract is an atypical location of metastatic lung cancer (0.5-1.3%), even though autopsy studies determine that its involvement is underdiagnosed (4.7-14%), especially when associated with the large cell subtype.1,3,5,6,8

The majority of patients are asymptomatic or present with the nonspecific symptoms of anemia, perforation, malabsorption, obstruction, or even appendicitis.3,5 Gastric or jejunal involvement can begin as a perforation and degenerate into secondary peritonitis, whereas tumors that grow rapidly usually begin as obstruction.3,5 Perforation is also related to the use of chemotherapy (especially bevacizumab) due to the tumor response secondary to treatment that produces cellular necrosis and thinning of the wall, favoring that complication.1

There is diagnostic suspicion of lung cancer in patients that begin with digestive symptoms or anemia, but those symptoms can also be the onset of the disease, given that they have been reported as the first manifestation in some case series. Gastrointestinal metastases condition a worse general outcome because they are related to a more advanced phase of disease when diagnosed.

Regarding perforation as a form of presentation, in 2004 Garwood carried out a review of 98 patients that presented with intestinal perforation secondary to metastatic lung cancer, identifying the jejunum as the first location (53%), followed by the ileum (28%). Perforation was associated with the histologic subtype of pulmonary adenocarcinoma (23.7%), followed by squamous cell carcinoma (22.7%), large cell carcinoma (20.6%), and small cell carcinoma (19.6%). Mean survival in this subgroup of patients was 66 days, 50% of the patients did not survive more than 30 days, and 1-year survival was less than 3%, 9,10

The majority of authors describe the small bowel as the main location of gastrointestinal involvement in 2.6-10.7% of the cases.2,3,5-9 The case series by Antler et al. is the exception. They describe the esophagus as the most affected organ (chiefly as a result of contiguity).6

Few cases of duodenal involvement are reported (15 cases published from 1999 to the present) and presentation is varied.

In 2011, Yamada et al. conducted a systematic bibliographic search of the cases of duodenal involvement published between 1999 and 2007, including the terms...