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ORIGINAL ARTICLE

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Pyloric atresia: report of two cases (one associated with epidermolysis bullosa and one associated with multiple intestinal atresias)

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Abstract We describe the US findings in two vomiting newborns affected by different forms of pyloric atresia, a rare congenital anomaly that includes a spectrum of lesions limited to the antro-pyloric region of the stomach and with various inheritance mechanisms and syndromic associations.

Keywords Pylorus · Atresia · Ultrasound

Introduction

Ultrasonography with high-resolution high-frequency equipment has become important in the evaluation of the paediatric gastrointestinal tract and is being used in an increasing number of applications. Although plain abdominal films can easily detect the presence of high intestinal obstruction, in non-bilious vomiting newborns, US is the easiest method to rule out or identify causes of gastric outlet obstruction.

Pyloric atresia is a rare congenital lesion that causes partial or complete obliteration of the gastric lumen. Although its precise incidence is unknown, it is believed to constitute less then 1% of all bowel atresias, giving it an estimated incidence of about 1 in 100,000 live births [1, 2].

We describe the US findings in two vomiting newborns affected by different forms of pyloric atresia, a rare congenital anomaly that includes a spectrum of lesions limited to the antro-pyloric region of the stomach and with various inheritance mechanisms and syndromic associations.

Case reports

Case 1

A baby girl was born after 34 weeks of gestation and an uneventful pregnancy. Physical examination at birth revealed abdominal distension and blistering skin lesions induced even with minor friction. Projectile bile-free vomiting appeared with the first oral feed. Inspection of the perineal region revealed an absent anus. Plain abdominal radiographs showed gastric dilatation with no air distal to the pylorus (Fig. 1a). Ultrasonography confirmed the distended stomach and demonstrated an abnormally elongated pylorus with thin single-layered walls, a normal duodenum and no evidence of compressing masses in the antral region (Fig. 1b, c). At laparoto-

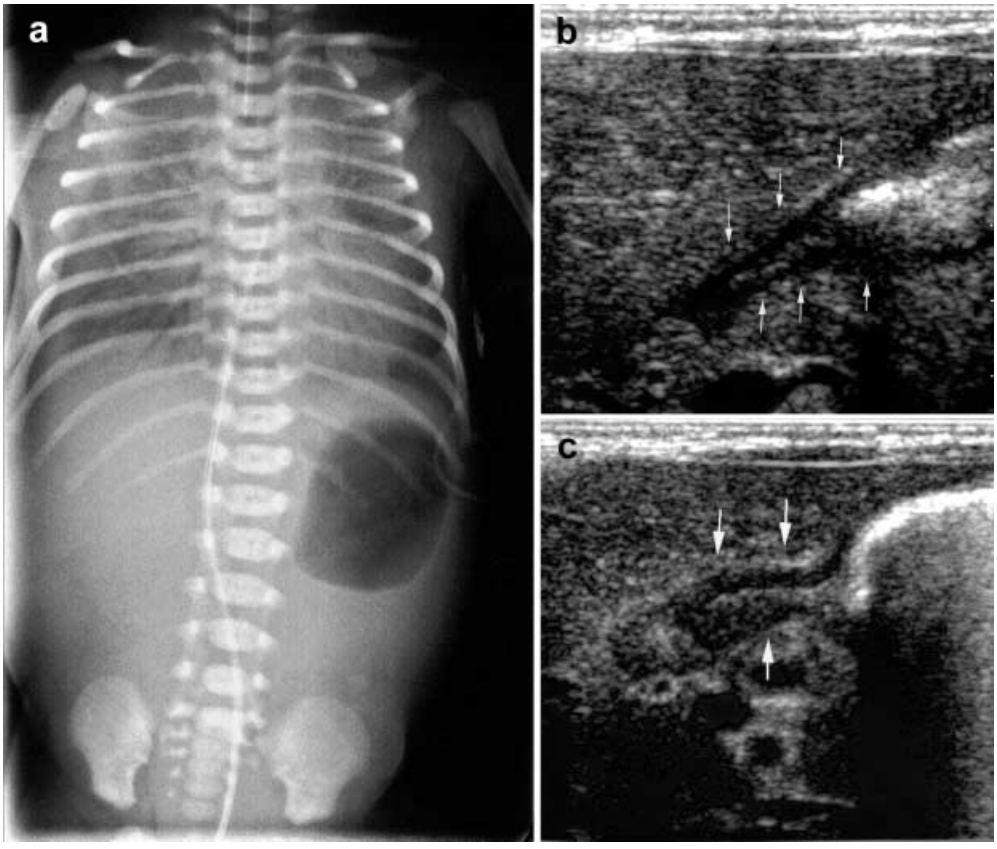

Fig. 1a–c. Case 1. **a** Plain thoraco-abdominal radiograph at birth reveals the single-bubble sign of gastric outlet obstruction and no air in the bowel loops distal to the pylorus. **b** Longitudinal and **c** oblique US images of the distal antrum show an abnormally elongated pylorus (**b**, *arrows*) with a thin single-layered wall (**c**, *arrows*) adjacent to the dilated gastric antrum. In **b**, the image has a 'tennis-racket'-like appearance. **c** The surgical specimen demonstrates a solid cord of tissue connecting the antrum to the duodenum

my, a solid cord connected the stomach to the duodenal bulb and gastroduodenostomy was performed (Fig. 1d). Histopathological examination of a skin biopsy, confirmed by electron microscopy and immunofluorescence techniques, was consistent with junctional epidermolysis bullosa. The baby died after a few days because of severe metabolic derangement.

Case 2

A female infant was delivered at the 32nd week of gestation by spontaneous vaginal delivery. History of pregnancy was not available as the mother was a migrant. Shortly after birth, the baby began non-bilious vomiting that was especially pronounced after feeding. Abdominal radiographs showed a large air-filled stomach, no air in the proximal and distal bowel, and scattered calcifications in the mid-abdomen consistent with calcified intraluminal meconium in the jejunal loops (Fig. 2a). Ultrasonography revealed a dilated stomach containing air and fluid material, and a large pseudocystic mass in the mid abdomen with a complex appearance (Fig. 2b). There were no other abnormalities. After 36 h, no meconium had been passed and the baby was taken to theatre. A completely obstructing antral gastric membrane, distal duodenal atresia associated with multiple jejunal and ileal atresias, and malrotation with the caecum in the left lower quadrant were found. She died soon after the operation and a congenital cardiac anomaly was identified at autopsy.

Discussion

Pyloric atresia, similar to other bowel atresias, is classified into three different anatomical varieties: type A is a prepyloric membrane, type B a solid core of tissue replacing the pyloric canal, and type C an atretic pylorus with a gap between stomach and duodenum [1, 2]. Based on 140 documented cases, the relative incidence of these variants is 57% for type A, 34% for type B and 9% for type C [3]. The two cases we describe belong respectively to type B and type A, the first with the surgical finding of a blind solid cord connecting the stomach and duodenum and the second with the gastric lumen completely obliterated by a thick mucosal membrane.

Pyloric atresia may occur as an isolated condition or associated with other abnormalities, the most common being junctional epidermolysis bullosa (EB), a rare autosomal recessive disorder affecting the skin and mucosa. It encompasses a group of blistering lesions that are classified into subgroups, depending on the precise ultrastructural level at which the split responsible for blistering occurs. Moreover, different types are associated with different outcomes; the junctional variety is the most severe and is often lethal [4, 5]. The association between pyloric atresia and EB, as in our case 1, is believed to be caused by the pleiotropic expression of a single gene because of the occurrence of these two rare congenital diseases in affected families [5]. Because EB can affect the mucosa of the gastrointestinal tract with subsequent inflammation and scarring, the majority of investigators have speculated that in these patients

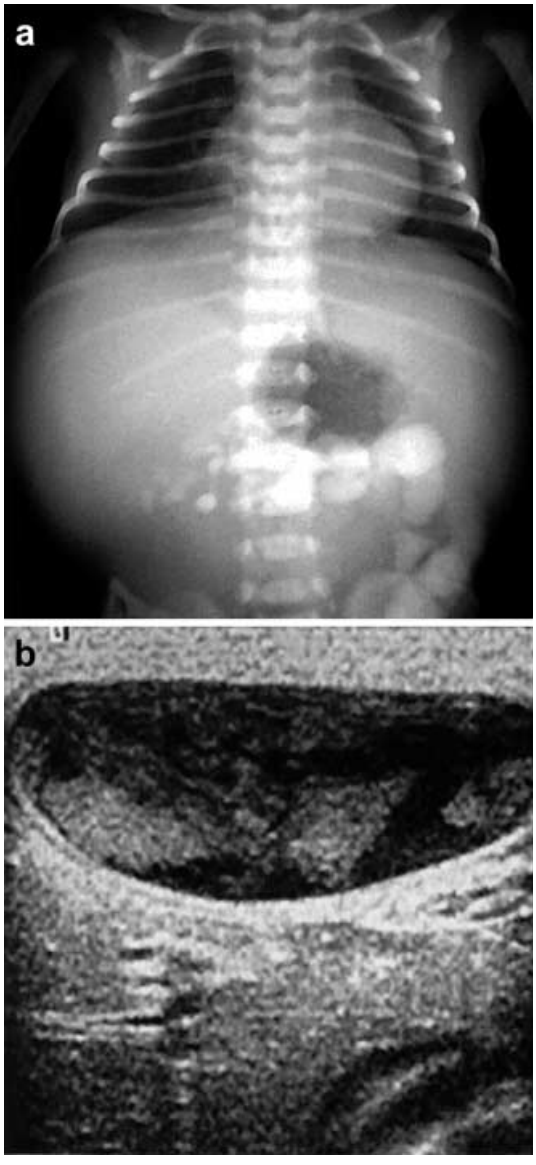

Fig. 2a, b. Case 2. **a** Plain abdominal radiograph displays signs of gastric outlet obstruction similar to case 1. Calcifications are visible in the mid and lower abdomen consistent with calcified intraluminal meconium. **b** Transverse US through the mid abdomen reveals a large pseudocystic mass with complex echoes consistent with meconium-filled dilated jejunum

pyloric atresia may result from an in utero-mucosal insult with subsequent luminal obliteration. However, scarring and inflammatory changes in the pyloric specimens have not been invariably demonstrated in affected newborns [2]. Nevertheless, the structure of $\alpha 6\beta 4$ integrin, a transmembrane glycoprotein that is engaged in cellular adhesion, migration and differentiation is known to be abnormal [6].

Pyloric atresia may also manifest as an isolated anomaly, or be associated with multiple intestinal atresias as in case 2. This association is familial and extremely rare; Al-Salem [1] recorded ten cases reported in the literature until 1999, and we found just one case in which malrotation and multiple atresias were described in the same newborn [7]. The syndrome of multiple intestinal atresias from the stomach to the rectum, together with calcification of intraluminal content between the atretic segments, is transmitted in an autosomal recessive pattern [8], with atresias typically occurring as type I or type II. Atretic lesions are believed to occur during the first trimester of pregnancy; they may also be associated with biliary dilatation, gall bladder agenesis and genitourinary and heart abnormalities, the latter being present in our case 2 [3, 9]. The mechanism by which gastrointestinal atresias result in these instances is not, so far, fully clarified, but is thought to be the consequence of a primary vascular accident or volvulus during intrauterine life.

The diagnosis of pyloric atresia may be suspected from plain abdominal films consistent with gastric outlet obstruction. A newborn with non-bilious vomiting, usually born to a mother with polyhydramnios, and presenting with radiographic evidence of a dilated stomach with the typical 'single bubble' appearance and no air beyond the pylorus has to be considered potentially affected by pyloric atresia. Lepinard et al. [10] described the prenatal diagnosis of pyloric atresia-junctional epidermolysis bullosa syndrome where the US findings were a dilated stomach, thin skin adherent to narrow nostrils and echogenic particles in the amniotic fluid. The differential diagnosis includes aberrant pancreatic tissue in the gastric antrum, antral stenosis and antral duplication cyst. Plain abdominal radiographs cannot assess the exact level and cause of obstruction, and diagnosis may be overlooked owing to the rarity of the disease.

Upper gastrointestinal series is almost never indicated as further investigation in such instances. It does not provide additional information because air is an effective and safe contrast medium.

Ultrasonography is quick and not invasive and adds additional information by demonstrating either the abnormal sonographic pattern of the antro-pyloric region or the absence of other causes of gastric obstruction, as well as many associated abnormalities that may affect the intestinal loops. Therefore, US complements plain abdominal films and may enable a specific diagnosis to be made, but few references exist in the literature describing the US findings of pyloric atresia.

In the first case we report that use of a high-frequency transducer demonstrated a long, stretched-out layered structure at the site of the pylorus with thin mural layers and absence of both the echolucent image of the pyloric muscle and the hyperechoic mucosal layers. It had a 'tennis-racket'-like shape and was connected to the dilated and fluid-filled gastric antrum (Fig. 1b). In case

2, US did not differentiate the thick and complete antral membrane found at surgery from the gastric content and gastric antral wall; the pylorus was not detected. Nevertheless, US did demonstrate proximal jejunal dilatation as a complex pseudocyst-like appearance as well as the calcifications already shown by the abdominal radiographs at the corresponding site and consistent with calcified intraluminal meconium in atretic loops.

Congenital antral membranes have been mostly described as incomplete and non-obstructive, and may be diagnosed later in life beyond the neonatal period. When the membrane is partial, symptoms may be delayed and the diagnosis elusive. Historically, congenital partial antral membranes have been diagnosed by barium meal studies. Chew et al. [11] reported the sonographic findings of a congenital antral membrane in an 18-day-old vomiting boy in whom a pierced antral diaphragm was detected as an echogenic structure bridging the lesser and greater curves; the pylorus was normal.

In conclusion, the management of the vomiting newborn affected by a congenital gastrointestinal lesion may be complex and requires fast, safe and reliable diagnostic methods in order to achieve the most appropriate surgical approach. Although pyloric atresia is often associated with an adverse prognosis, as in the two cases we describe, and may lead to the opinion that surgical intervention should be withheld, an increasing number of long-term survivors has been reported [4, 5]. High-frequency US should be considered the diagnostic method of choice for the evaluation of the gastrointestinal tract. In the two cases we present, US was valuable because it directly visualised the abnormal pylorus or because it allowed evaluation of the anatomy of neighbouring organs and identified associated anomalies. The upper gastrointestinal series may be dangerous when there is the risk of inhalation risk and does not distinguish antro-pyloric congenital abnormalities from other causes of gastric outlet obstruction in the newborn.

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