Pyloric Atresia and Prepyloric Antral Diaphragm

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Abstract

Pyloric atresia (PA) and prepyloric antral diaphragm are two causes of congenital gastric outlet obstruction.

PA is a very rare condition, with an incidence of about 1 in 100,000 newborns, representing approximately 1% of all intestinal atresias. The exact etiology of this condition is not known. Commonly, PA is an isolated condition, but familial occurrence is also reported, supporting a genetic predisposition. Most of

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P. Puri (ed.), *Pediatric Surgery*,
https://doi.org/10.1007/978-3-662-43588-5_59

the familial cases have been reported to be associated with epidermolysis bullosa (EB-PA) and are characterized by an autosomal recessive inheritance. Prenatal diagnosis is possible on ultrasound (US); prenatal testing in families at risk for EB-PA can be performed. After birth, the main presenting symptoms are non-bilious vomiting and upper abdominal distension. Associated anomalies are very frequent, the commonest being EB. The diagnosis can be made on plain abdominal X-ray, barium meal, and abdominal US. The treatment is surgical and the prognosis is variable, mostly depending on the severity of the associated conditions.

Prepyloric antral diaphragm is a rare condition, whose prevalence and etiology are still unknown. The age of onset and clinical presentation vary depending on the degree of obstruction determined by the diaphragm. The main symptoms are recurrent non-bilious vomiting, often projectile; progressive feeding problems; epigastric pain; and failure to thrive. The diagnosis can be made on plain abdominal films, barium meal, and abdominal US; endoscopy is helpful and may also be a therapeutic tool. Surgery is the treatment of choice and the prognosis is excellent.

Keywords

Gastric outlet obstruction (GOO) · Pyloric atresia (PA) · Prepyloric antral diaphragm · Epidermolysis bullosa (EB) · Hereditary multiple intestinal atresias (HMIA) · Carmi syndrome · Integrin gene (*ITG*) mutations · Plectin (*PLEC*) gene mutations · Pyloroplasty · Gastroduodenostomy

Introduction

Pyloric atresia and prepyloric antral diaphragm are two causes of gastric outlet obstruction (GOO), a broad spectrum of conditions that prevent the passage of gastric contents into the duodenum.

GOO is a relatively common condition in pediatric age, if hypertrophic pyloric stenosis (HPS) is considered. However, when HPS is excluded, GOO is quite rare, with an incidence of 1 in 100,000 live births (Sharma et al. 2008).

The causes of GOO may be classified as follows (Sharma et al. 2008):

1. Congenital intrinsic obstruction of antrum and pylorus:
   (a) Aplasia
   (b) Atresia
   (c) Diaphragms and webs
   (d) Luminal obstruction (e.g., mucosal valves, heterotrophic pancreas, etc.)
2. HPS
3. Acquired:
   (a) Primary:
      (i) Acquired gastric outlet obstruction during infancy and childhood
   (b) Secondary:
      (i) Acid peptic disease (chronic duodenal or juxtapyloric ulcer)
      (ii) Neoplasm
      (iii) Chemical injury (ingestion of acid, caustic, and other irritants like potassium carbonate)
      (iv) Others: chronic granulomatous disease, Crohn's disease, eosinophilic gastritis, gastric duplication cysts, and cholecystogastrocolic band (malrotation) (Nissan et al. 1997)

Pyloric Atresia

Definition and Classification

Congenital pyloric atresia (PA) was firstly described by Calder in 1749. The first successful surgical correction was performed in 1940 by Touroff (Al-Salem 2007; Ilce et al. 2003).

Anatomically, it is divided into three types:

– Type 1, the commonest: pyloric membrane or web (57%)

– Type 2: pyloric atresia with a solid cord between the two ends (34%)
– Type 3: pyloric atresia with a gap between the stomach and the duodenum (9%) (Al-Salem 2007; Ilce et al. 2003; Tomá et al. 2002).

Epidemiology

Congenital PA is a very rare condition, with an incidence of about 1 in 100,000 newborns. It represents approximately 1% of all intestinal atresias (Al-Salem 2007; Ilce et al. 2003; Chung and Uitto 2010; Mboyo et al. 2016).

Sex distribution is about equal (Ilce et al. 2003; Andriessen et al. 2010).

Etiopathogenesis

The exact etiology of this condition is not known.

Embryologically, it is thought to result from a developmental arrest between the 5th and 12th week of intrauterine life. According to Tandler's theory, this anomaly is the result of a failure of the tube canalization during development (Al-Salem 2007; Andriessen et al. 2010; Sencan et al. 2002).

Commonly, PA is an isolated condition, but familial occurrence is also reported, supporting a genetic predisposition. Most of the familial cases have been reported to be associated with epidermolysis bullosa (EB) and are characterized by an autosomal recessive inheritance (Chung and Uitto 2010; Birnbaum et al. 2008; Charlesworth et al. 2013; Dang et al. 2008; Abe et al. 2007; Natsuga et al. 2010; Usui et al. 2013).

PA in association with EB is supposed to result from two pathologic elements (Al-Salem 2007; Ilce et al. 2003; Natsuga et al. 2010; Bıçakcı et al. 2012):

1. Intrauterine sloughing of the pyloric mucosa, as a result of disintegration of basement membrane and hemidesmosomes
2. Subsequent inflammatory response which causes fibrous cicatrization and obliteration of the intestinal lumen, especially in anatomically narrow passages, such as the pyloric canal.

Hereditary multiple intestinal atresias (HMIA), frequently associated with PA, are reported as an autosomal recessive condition, although the candidate gene has not been identified yet (Al-Salem 2007; Cole et al. 2010).

In patients affected by epidermolysis bullosa and pyloric atresia (EB-PA) and Carmi syndrome (EB-PA and aplasia cutis congenita), the affected skin presents morphological abnormalities in the hemidesmosomes and reduced or absent expression of the hemidesmosomal component integrin α6β4, a member of a large family of α/β heterodimeric transmembrane glycoprotein receptors (Chung and Uitto 2010; Birnbaum et al. 2008; Abe et al. 2007; Ozge et al. 2012).

Sequence analysis of integrin genes identified mutations in integrin β4, encoded by integrin β4 (*ITGB4)* gene, in most EB-PA cases, with few cases demonstrating integrin α6 (*ITGA6* gene) mutations (Chung and Uitto 2010; Birnbaum et al. 2008). Almost 70 distinct mutations of *ITGB4* in EB-PA patients can be found in the mutation database, while a total of 5 mutations in the *ITGA6* gene have been reported (Chung and Uitto 2010). Sequencing analysis shows that *ITGB4* accounts for 80% and *ITGA6* for 5% of EB-PA patients (Ozge et al. 2012).

ITGB4 mutation database suggests that premature termination codon (PTC) mutations predominantly result in lethal forms, while missense mutations frequently associate with nonlethal variants (Abe et al. 2007; Ozge et al. 2012).

Other genetic mutations that have been linked to EB-PA lie in the gene for plectin (*PLEC*) (Charlesworth et al. 2013; Nakamura et al. 2011; Natsuga 2015). Plectin is a cytoskeletal protein abundantly expressed in several cell types (epithelia, muscles, and fibroblasts). It harbors binding sites for several proteins, including integrin α6β4. The association of *PLEC* mutations with EB-PA is therefore consistent with plectin's cross-linking functions with other proteins and results in pleiotropic phenotypes (Charlesworth et al. 2013). Genetic findings suggest that EB-PA

is linked to mutations in the portions of plectin that mediate interactions with integrin β4 (Charlesworth et al. 2013). Ten distinct mutations in the *PLEC1* gene have been demonstrated in patients with EB-PA (Chung and Uitto 2010). EB-PA due to *PLEC* mutations is considered universally lethal (Nakamura et al. 2011).

A mutation in the *ITGB4* gene has been recently detected in a patient affected by PA and desquamative enteropathy, without skin disease (Salvestrini et al. 2008).

Diagnosis

Prenatal Diagnosis and Counselling

Prenatal diagnosis of PA is possible on ultrasound (US) (Al-Salem 2007; Usui et al. 2013), with visualization of polyhydramnios (reported in about 50% of PA cases (Ilce et al. 2003)), a dilated stomach in the absence of a double bubble (Andriessen et al. 2010), and narrowing of the gastric outlet (Ilce et al. 2003).

Cases of diagnostic confirmation by fetal magnetic resonance imaging have been reported (Yu et al. 2009).

The presence of EB or Carmi syndrome can be suggested prenatally by the sonographic so-called "snowflake" sign (echogenic particles in the amniotic fluid), associated with fetal stomach dilation and polyhydramnios (Birnbaum et al. 2008; Usui et al. 2013).

EB can be also diagnosed by electron microscopy or immunofluorescence of a fetal skin biopsy (Al-Salem 2007; D'Alessio et al. 2008).

Identification of genetic mutations in families has provided molecular confirmation of diagnosis together with prognostication and can also provide a means for prenatal testing in families at risk for recurrence of the disease (Chung and Uitto 2010; Nakamura et al. 2011).

Prenatal testing in families at risk for EB-PA can be performed from chorionic villus sampling, which can be performed as early as the 10th week of gestation (Chung and Uitto 2010) or from amniotic fluid sampling (Nakamura et al. 2011).

A new technique of immunofluorescence analyses of villous trophoblastus has been recently proposed (D'Alessio et al. 2008).

Preimplantation genetic diagnosis, for couples at risk of having children with EB-PA, has been also offered to avoid abortion of possibly affected pregnancies (Ozge et al. 2012).

The prognosis, the decision about whether to continue or not the pregnancy, and the risks for future pregnancies should be discussed with the parents, advising them about the risk of recurrence (25% for autosomal recessive diseases) and the prognosis associated with different types of mutations, together with the encouraging improvements of the medical therapy to treat EB (Al-Salem 2007; Parshotam et al. 2007).

Clinical Presentation

Almost all patients have a low weight at birth (Ilce et al. 2003).

After birth, the main presenting symptoms are non-bilious vomiting and upper abdominal distension (Al-Salem 2007; Andriessen et al. 2010).

Congenital PA can be an isolated condition, but associated anomalies are also very frequent (30–50% of cases). The commonest associated anomaly is EB (Al-Salem 2007).

EB is categorized in three major groups: EB simplex (EBS), junctional EB (JEB), dystrophic EB (DEB), and Kindler syndrome, depending on the depth of the dermal-epidermal junction split (Nakamura et al. 2011). EB-PA is classified as a form of JEB (Ozge et al. 2012).

Skin lesions may not appear until as late as 48 h after birth. They are associated with disruption of the intestinal mucosa, which causes malabsorption, increased antigenic sensitivity, bloody diarrhea, and protein losing enteropathy (Bıçakcı et al. 2012).

The severity of skin involvement in EB-PA can be variable, and both lethal and nonlethal variants have been reported (Chung and Uitto 2010; Abe et al. 2007). In some patients, even on successful surgical correction of the pyloric or intestinal atresia, skin involvement is so severe that the children die from complications, such as infections or electrolyte imbalance, within a few days or weeks from birth. In others, skin fragility can be mild,

or it can improve with age, allowing them to conduct normal lives (Chung and Uitto 2010).

EB-PA can be associated to many other disorders including gastrointestinal, urinary, pulmonary, and eye problems, as well as dental (enamel hypoplasia, dental caries), hair (alopecia), and nail (toenail dystrophy) abnormalities, ear or nose hypoplasia, or atrophy (Dang et al. 2008; Abe et al. 2007; Bıçakcı et al. 2012). The associated anomaly rate is reported to be around 30% (Ilce et al. 2003).

The combination of PA, EB, and aplasia cutis congenita (PA-EB-ACC) is known as Carmi syndrome (OMIM #226730) (Birnbaum et al. 2008). It is characterized by congenital absence of skin, mostly of the limbs, involving all skin layers and severe neonatal mucocutaneous blistering (Chung and Uitto 2010; Birnbaum et al. 2008). Most cases result in early death of the affected child, but nonlethal cases with progressive improvement of the cutaneous lesions have been described (Birnbaum et al. 2008).

In 2010 the first report of EB complicated with both PA and muscular dystrophy was published, caused by mutations in the plectin gene (*PLEC*) (Natsuga et al. 2010).

PA can also be associated with distal duodenal atresia forming a closed duodenal loop, where biliary and pancreatic secretions accumulate, leading to massive distention and possible duodenal perforation (Al-Salem 2007), esophageal atresia (Al-Salem 2007; Ilce et al. 2003), hereditary multiple intestinal atresias (HMIA), with or without combined immunodeficiency syndrome (Cole et al. 2010; Bass 2002), intrauterine growth retardation, Down's syndrome, congenital heart disease, cleft palate, Meckel's diverticulum (Al-Salem 2007), Dieulafoy lesion of the stomach (Polonkai et al. 2011), ureterovesical junction obstruction, pelviureteric junction obstruction, agenesis of the gallbladder (Al-Salem 2007), ectopic drainage of the common bile duct, annular pancreas (Scheida et al. 2009), malrotation, anorectal agenesis (Al-Salem 2007), and pylorocholedochal fistula (Al-Salem 2007; Sencan et al. 2002).

Laboratory

During the prenatal period, elevated levels of maternal serum and/or amniotic fluid alpha-fetoprotein can be found in EB-PA (Birnbaum et al. 2008; Yu et al. 2009).

In patients with EB-PA, immunofluorescence mapping of the affected skin usually shows completely negative staining for integrin β4 and/or integrin α6 in lethal cases, whereas nonlethal forms are usually associated with positive but attenuated staining (Dang et al. 2008).

Radiologic Findings

The diagnosis can be made on plain abdominal X-ray, which shows a single, large air bubble, representing the dilated stomach, with no gas distally (Al-Salem 2007; Parshotam et al. 2007; Hermanowicz and Debek 2015).

A double bubble may, however, be seen in cases with prolapse of a pyloric membrane into the duodenum, thus imitating duodenal atresia. An intermittent double bubble sign has also been described, probably reflecting configuration of the distended stomach (Parshotam et al. 2007).

The presence of calcification raises the possibility of associated multiple intestinal atresias (Al-Salem 2007): they are homogeneous intraluminal calcifications, described as a "string of pearls," almost resembling a contrast study, and are pathognomonic of this condition (Bass 2002).

Barium meal can confirm the diagnosis, as it shows dilated stomach with obstruction at the pylorus level and no contrast passing distally (Al-Salem 2007).

An additional lower gastrointestinal contrast study (barium enema) can be done to rule out the presence of colonic atresia (Al-Salem 2007).

US with high-resolution high-frequency equipment is a useful and noninvasive technique in the evaluation of PA and associated anomalies, demonstrating the abnormal sonographic pattern of the antropyloric region, the absence of other causes of gastric obstruction, as well as many associated anomalies that may affect the intestinal loops (distal loops' dilatation in case of multiple atresias, calcifications) or other abdominal organs (Tomá et al. 2002).

Abdominal US can show bile duct dilatation and cystic dilation of the duodenum in case of both PA and duodenal or high jejunal obstruction with retention of pancreaticobiliary secretions (Bass 2002).

Differential Diagnosis

Non-bilious vomiting in the newborn period can be interpreted as gastroesophageal reflux, the most frequent cause of delayed emptying in children, leading to a delay in the diagnosis of PA (Ilce et al. 2003; Polonkai et al. 2011).

Other congenital developmental anomalies, such as hypertrophic pyloric stenosis, pyloric duplication, antrum membrane, gastric volvulus, gastric duplication, aberrant pancreatic tissue plugging the pylorus, and retrograde duodenogastric intussusception, need to be included in the differential diagnosis of PA (Ilce et al. 2003; Tomá et al. 2002; Polonkai et al. 2011).

Treatment

The treatment of PA is surgical (Fontenot et al. 2011).

Preoperative management should include nasogastric tube placement, intravenous fluid administration, and electrolyte correction (Andriessen et al. 2010; Fontenot et al. 2011).

Intraoperatively, it is important to make sure that only one pyloric diaphragm is present, as this can be multiple (Al-Salem 2007; Zecca et al. 2010). It is also important to check the patency of the distal intestine, passing a catheter and injecting saline distally to exclude associated intestinal atresias.

The surgical treatment depends on the type of PA discovered at laparotomy (Al-Salem 2007; Fontenot et al. 2011).

Type 1 is treated by excision of the membrane/web with or without Heineke-Mikulicz or Finney pyloroplasty (Al-Salem 2007; Ilce et al. 2003; Dessanti et al. 2004; Yokoyama and Utsunomiya 2012). Recent advances in endoscopy have allowed the development of alternative treatment strategies, including balloon dilatation, laser web excision, and laser radial incision (Yokoyama and Utsunomiya 2012).

For type 2, excision of the atretic segment with end-to-end or diamond-shaped (proximal transverse and distal longitudinal incision, Kimura technique) gastroduodenostomy is recommended as the standard technique (Andriessen et al. 2010; Dessanti et al. 2004).

A different procedure has been also proposed, the *gastroduodenal mucosal advancement anastomosis*, which achieves an anatomic reconstruction of the pyloric canal (Dessanti et al. 2004). The atretic pylorus is incised longitudinally, showing the presence of an internal muscular layer, which is separated as in Ramstedt operation. By this pyloromyotomy, the mucosal cul-de-sacs on the gastric and duodenal sides are isolated, mobilized, and advanced into the pyloric canal and then sutured into an end-to-end anastomosis. The pyloromyotomy is then closed.

A novel gastroduodenostomy procedure for solid segment type has been recently published (Yokoyama and Utsunomiya 2012), through which the stomach and duodenal lumen are anastomized end to end anteriorly to the solid segment, which is not excised.

When a gap is present (type 3), the treatment is gastroduodenostomy (Andriessen et al. 2010; Fontenot et al. 2011).

Gastrojejunostomy should be avoided because it is associated with high failure and mortality rates (Ilce et al. 2003; Yokoyama and Utsunomiya 2012).

When PA is associated with EB, surgical intervention is often effective only for short-term survival, because infants usually die within the first few weeks or months of life, due to complications related to EB (Bıçakcı et al. 2012).

Identification of the genetic mutations involved in EB-PA provides the basis for molecular therapies: gene therapy, protein replacement or stem cell therapies. These approaches are supported by the development of animal models of different forms of EB, including those with targeted ablation of the *ITGB4*, *ITGA6*, and *PLEC1* genes (Chung and Uitto 2010).

Some forms of desquamative enteropathies associated with PA have been treated with immune modulatory therapy with significant clinical improvement (Salvestrini et al. 2008).

Complications

After excision of the atretic pylorus and gastroduodenostomy, pyloric function can be lost with consequent duodenogastric reflux, which can cause gastric disorders like alkaline gastritis as a result of bilious reflux into the stomach (Yokoyama and Utsunomiya 2012).

Outcome

The prognosis of PA is variable.

Isolated PA, particularly if type 1, if diagnosed and treated on time, has an excellent prognosis (Ilce et al. 2003; Fontenot et al. 2011). Mortality rate is higher if there is a diagnostic delay (Ilce et al. 2003) and if atresia contains a solid component (Fontenot et al. 2011).

The presence of associated anomalies is an important factor for the reported high mortality rate, exceeding 50% (Yu et al. 2009; Fontenot et al. 2011).

EB is generally fatal because of fluid and protein loss, electrolyte imbalance and sepsis (Al-Salem 2007; Bıçakcı et al. 2012). This is why some surgeons think that the association of congenital PA and EB should preclude surgical treatment of PA. However, recent advances in the medical treatment of EB, with the use of steroids and phenytoin, have been reported to give favorable results (Parshotam et al. 2007).

Congenital PA in association with hereditary multiple intestinal atresias (HMIA) is reported as always fatal, the cause of death being sepsis in the majority of cases (Al-Salem 2007; Cole et al. 2010), particularly when HMIA is associated with immunodeficiency (Cole et al. 2010; Bass 2002). Awareness of the pathognomonic findings of HMIA in plain abdominal X-ray and US must alert the surgeon of the severity of this condition, because it might alter medical and surgical management (Bass 2002).

Prepyloric Antral Diaphragm

Syn.: **(congenital) (gastric) antral (mucosal) web/membrane/diaphragm – prepyloric (mucosal) web/diaphragm**

Definition

Prepyloric antral membrane (PAM) is a rare cause of congenital gastric outlet obstruction (Borgnon et al. 2003), firstly described in 1933 by Parsons and Barling (Parsons and Barling 1933).

It is a circumferential intraluminal mucosal septum in the prepyloric region, perpendicular to the long axis of the antrum (Chao et al. 2011). It consists of normal, noninflamed mucosal and submucosal gastric mural layers (Otjen et al. 2012) which may or may not have a muscular component (de Vries et al. 2011; Ferguson et al. 2004). The antral web is usually 2–4 mm thick, located 1–7 cm proximal to the pyloroduodenal junction (Chao et al. 2011; Otjen et al. 2012; Tiao et al. 2005). In most of cases, it has a central or eccentric perforation, which may range from 2 to 30 mm in diameter; in a few cases, the web is unperforated or complete (Chao et al. 2011).

Epidemiology

This is a rare condition, whose prevalence is unknown; only sporadic cases are described in the literature.

Etiopathogenesis

The etiology is unknown.

PAM is considered a congenital lesion resulting from a local redundancy of the endodermal tube (Borgnon et al. 2003).

A report published in 1997 (Gahukamble 1998) describes multiple occurrence of this condition in a close family unit, suggesting the hypothesis of a genetic etiology, with autosomal recessive inheritance being the most probable mode of transmission.

Diagnosis

Clinical Presentation

This condition has been described in different ages, from premature and newborn infants to adults (Chao et al. 2011). The age of onset and clinical presentation vary depending on the degree of obstruction determined by the diaphragm – symptoms usually correlating with apertures of 1 cm or less (Otjen et al. 2012; Tiao et al. 2005), with the consistency of ingested food and with the ability of the gastric motility to overcome the partial luminal web (Chao et al. 2011; de Vries et al. 2011).

A complete web presents with persistent vomiting commencing shortly after birth, while incomplete antral membranes are usually diagnosed at an older age (3–11 years) and exceptionally in adults (Nissan et al. 1997; de Vries et al. 2011; Lui et al. 2000).

The main symptoms are recurrent non-bilious vomiting (Borgnon et al. 2003), often projectile (Noel et al. 2000), progressive feeding problems particularly for solid nutrition, epigastric pain (sometimes presenting in infants with unexplained excessive crying (Noel et al. 2000)), and failure to thrive (de Vries et al. 2011; Feng et al. 2005). Cases are described who became symptomatic after foreign body ingestion (Oak et al. 1996).

Some authors described misleading presentation of prepyloric web with prominent pulmonary symptoms due to secondary gastroesophageal reflux with probable recurrent aspiration pneumonias (de Vries et al. 2011).

Antral webs associated with hypertrophic pyloric stenosis, distal antral hypertrophy, or duodenal atresia have been described (Ferguson et al. 2004; Tiao et al. 2005).

Radiologic Findings

Plain abdominal films show a distended gas-filled stomach (Borgnon et al. 2003).

Ultrasound shows a fluid-filled, dilated stomach (Chao et al. 2011), with very limited passage of gastric content from the stomach to the duodenum, active gastric peristalsis but no increase of the pyloric wall thickness or canal length (Borgnon et al. 2003; de Vries et al. 2011); Chew et al. (Chew et al. 1992) proposed four ultrasound diagnostic criteria of an antral web, including demonstration of an echogenic diaphragm-like structure in the antral region, gastric dilatation, delay in gastric emptying, and a normal pylorus.

A diagnosis can be established through a barium meal study in most of cases (Chao et al. 2011; de Vries et al. 2011), with a 90% sensitivity reported (Lui et al. 2000). Upper gastrointestinal series demonstrate a dilated stomach with delayed gastric emptying, sometimes revealing a transverse radiolucent line in the antral region (Borgnon et al. 2003) as well as a spraying of barium through a central or an eccentric aperture with "jet stream" during examination (Chao et al. 2011; de Vries et al. 2011). A "double bulb" sign has been described when a thicker membrane mimics the pylorus, creating an antral chamber (Otjen et al. 2012).

Endoscopy is helpful in the diagnosis of PAM, in the detection of other gastric pathologies (e.g., peptic diseases, adhesions) (Nissan et al. 1997; de Vries et al. 2011), and may also be a therapeutic tool (Borgnon et al. 2003).

However, in many cases, the definite diagnosis is achieved only during surgical exploration (Borgnon et al. 2003).

Differential Diagnosis

The main entities in the differential diagnosis are pyloric stenosis, pylorospasm, redundant gastric mucosal folds, perigastric adhesions, heterotopic pancreas tissue (Borgnon et al. 2003; Lui et al. 2000), irregular webs, and deformity of the antrus suggesting acquired etiologies such as ulcer (Lui et al. 2000) and gastroesophageal reflux (de Vries et al. 2011).

One case of intramuscular circumferential prepyloric fibrous band in the gastric wall causing partial obstruction and mimicking antral web has recently been described (Medsinge et al. 2012).

Symptoms may mimic duodenal ulcer (Nissan et al. 1997).

Treatment

For antral diaphragm with gastric outlet obstruction, surgery is the treatment of choice (Chao et al. 2011; de Vries et al. 2011). It consists of incision or excision of the membrane by gastrostomy (de Vries et al. 2011; Ferguson et al. 2004), with or without associated pyloroplasty (Borgnon et al. 2003; Ferguson et al. 2004; Gahukamble 1998; Lui et al. 2000).

An endoscopic balloon dilatation or transection of the web can be performed (Borgnon et al. 2003; Lui et al. 2000; Feng et al. 2005).

Complications

A reported complication of the surgical correction is pyloric scarring with subsequent pyloric stricture, which has been treated through endoscopic procedures (Chao et al. 2011).

Outcome

The prognosis is excellent (Nissan et al. 1997).

Persistence of minimal feeding problems because of the secondary atonic stomach can occur, particularly in case of late diagnosis and treatment (de Vries et al. 2011).

Conclusion and Future Directions

Pyloric atresia and prepyloric antral diaphragm are rare conditions, but they are important to keep in mind when evaluating a patient with clinical symptoms suggesting gastric outlet obstruction, because of their potential unfavorable prognosis.

Barium meal and abdominal ultrasound are the most important tools in diagnosing these entities. Because each condition has a different imaging appearance and diagnosis can be challenging, experience with the use of these modalities and a working differential diagnosis are important for a timely diagnosis.

While prepyloric antral diaphragm usually has a less severe clinical presentation and a better outcome, PA can be associated with a variety of associated conditions, particularly skin lesions, that can significantly increase morbidity and mortality. Nevertheless, an early diagnosis can often improve the outcome, and a better insight into the genetics and the immunologic aspects of this condition will probably provide the basis for a more structured and widespread use of prenatal diagnosis, for the implementation of immunomodulatory therapies, and for the development of molecular therapies.

Cross-References

- ▶ Anatomy of the Infant and Child
- ▶ Colonic and Rectal Atresias
- ▶ Duodenal Obstruction
- ▶ Embryology of Congenital Malformations
- ▶ Fetal Counseling for Congenital Malformations
- ▶ Fluid and Electrolyte Balance in Infants and Children
- ▶ Infantile Hypertrophic Pyloric Stenosis
- ▶ Jejunoileal Atresia and Stenosis
- ▶ Prenatal Diagnosis of Congenital Malformations
- ▶ Principles of Pediatric Surgical Imaging
- ▶ Sepsis

References

Abe M, Sawamura D, Goto M, Nakamura H, Nagasaki A, Nomura Y, Kawasaki H, Isogai R, Shimizu H. ITGB4 missense mutation in a transmembrane domain causes non-lethal variant of junctional epidermolysis bullosa with pyloric atresia. J Dermatol Sci. 2007;47(2):165–7.

Al-Salem AH. Congenital pyloric atresia and associated anomalies. Pediatr Surg Int. 2007;23(6):559–63.

Andriessen MJ, Matthyssens LE, Heij HA. Pyloric atresia. J Pediatr Surg. 2010;45(12):2470–2.

Bass J. Pyloric atresia associated with multiple intestinal atresias and immune difficiency. J Pediatr Surg. 2002;37(6):941–2.

Bıçakcı U, Tander B, Cakmak Çelik F, Arıtürk E, Rızalar R. Pyloric atresia associated with epidermolysis bullosa: report of two cases and review of the literature. Ulus Travma Acil Cerrahi Derg. 2012;18(3):271–3.

Birnbaum RY, Landau D, Elbedour K, Ofir R, Birk OS, Carmi R. Deletion of the first pair of fibronectin type III repeats of the integrin beta-4 gene is associated with epidermolysis bullosa, pyloric atresia and aplasia cutis congenita in the original Carmi syndrome patients. Am J Med Genet A. 2008;146A(8):1063–6.

Borgnon J, Ouillon-Villet C, Huet F, Sapin E. Gastric outlet obstruction by an antral mucosal diaphragm: a case of a congenital anomaly revealed by an acquired disease. Eur J Pediatr Surg. 2003;13(5):327–9.

Chao HC, Luo CC, Wang CJ. Elimination of postoperative pyloric stricture by endoscopic electrocauterization and balloon dilatation in an infant with congenital antral web. Pediatr Neonatol. 2011;52(2):106–9.

Charlesworth A, Chiaverini C, Chevrant-Breton J, Delrio M, Diociaiuti A, Dupuis RP, El Hachem M, Le Fiblec B, Sankari-Ho AM, Valhquist A, Wierzbicka E, Lacour JP, Meneguzzi G. Epidermolysis bullosa simplex with PLEC mutations: new phenotypes and new mutations. Br J Dermatol. 2013;168(4):808–14.

Chew AL, Friedwald JP, Donovan C. Diagnosis of congenital antral web by ultrasound. Pediatr Radiol. 1992;22:342–3.

Chung HJ, Uitto J. Epidermolysis bullosa with pyloric atresia. Dermatol Clin. 2010;28(1):43–54.

Cole C, Freitas A, Clifton MS, Durham MM. Hereditary multiple intestinal atresias: 2 new cases and review of the literature. J Pediatr Surg. 2010;45(4):E21–4.

D'Alessio M, Zambruno G, Charlesworth A, Lacour JP, Meneguzzi G. Immunofluorescence analysis of villous trophoblasts: a tool for prenatal diagnosis of inherited epidermolysis bullosa with pyloric atresia. J Invest Dermatol. 2008;128(12):2815–9.

Dang N, Klingberg S, Rubin AI, Edwards M, Borelli S, Relic J, Marr P, Tran K, Turner A, Smith N, Murrell DF. Differential expression of pyloric atresia in junctional epidermolysis bullosa with ITGB4 mutations suggests that pyloric atresia is due to factors other than the mutations and not predictive of a poor outcome: three novel mutations and a review of the literature. Acta Derm Venereol. 2008;88(5):438–48.

Dessanti A, Di Benedetto V, Iannuccelli M, Balata A, Cossu Rocca P, Di Benedetto A. Pyloric atresia: a new operation to reconstruct the pyloric sphincter. J Pediatr Surg. 2004;39(3):297–301.

de Vries AG, Bodewes FA, van Baren R, Karrenbeld A, Broens PM. Misleading clinical symptoms and a prolonged diagnostic approach in prepyloric web. J Pediatr Gastroenterol Nutr. 2011;52(5):627–9.

Feng J, Gu W, Li M, Yuan J, Weng Y, Wei M, Zhou X. Rare causes of gastric outlet obstruction in children. Pediatr Surg Int. 2005;21(8):635–40.

Ferguson C, Morabito A, Bianchi A. Duodenal atresia and gastric antral web. A significant lesson to learn. Eur J Pediatr Surg. 2004;14(2):120–2.

Fontenot B, Streck C, Hebra A, Cina R, Gutierrez P. Pyloric atresia. Am Surg. 2011;77(2):249–50.

Gahukamble DB. Familial occurrence of congenital incomplete prepyloric mucosal diaphragm. J Med Genet. 1998;35(12):1040–2.

Hermanowicz A, Debek W. Images in clinical medicine. Single bubble – pyloric atresia. N Engl J Med. 2015;373(9):863.

Ilce Z, Erdogan E, Kara C, Celayir S, Sarimurat N, Senyüz OF, Yeker D. Pyloric atresia: 15-year review from a single institution. J Pediatr Surg. 2003;38(11):1581–4.

Lui KW, Wong HF, Wan YL, Hung CF, Ng KK, Tseng JH. Antral web – a rare cause of vomiting in children. Pediatr Surg Int. 2000;16(5–6):424–5.

Mboyo A, Clermidi P, Podevin G. Neonatal gastric outlet obstruction by isolated pyloric atresia, an often forgotten diagnosis. Acta Chir Belg. 2016;116(2):89–95.

Medsinge A, Bartoletti SC, Furtado A, Katz A, Tadros SS. Isolated intramuscular fibrous band masquerading antral web in a premature neonate. A case report. J Pediatr Surg. 2012;47(8):e17–9.

Nakamura H, Natsuga K, Nishie W, McMillan JR, Nakamura H, Sawamura D, Akiyama M, Shimizu H. DNA-based prenatal diagnosis of plectin-deficient epidermolysis bullosa simplex associated with pyloric atresia. Int J Dermatol. 2011;50(4):439–42.

Natsuga K. Plectin-related skin diseases. J Dermatol Sci. 2015;77(3):139–45.

Natsuga K, Nishie W, Shinkuma S, Arita K, Nakamura H, Ohyama M, Osaka H, Kambara T, Hirako Y, Shimizu H. Plectin deficiency leads to both muscular dystrophy and pyloric atresia in epidermolysis bullosa simplex. Hum Mutat. 2010;31(10):E1687–98.

Nissan A, Seror D, Udassin R. Gastric outlet obstruction caused by prepyloric mucosal diaphragm mimicking duodenal ulcer: a case report. Acta Paediatr. 1997;86 (1):116–8.

Noel RJ, Glock MS, Pranikoff T, Hill ID. Nonobstructive antral web: an unusual cause of excessive crying in an infant. J Pediatr Gastroenterol Nutr. 2000;31 (4):439–41.

Oak S, Bhatnagar M, Kulkarni B, Nanivadekar S, Karmarkar S, Sawant P. Prepyloric diaphragm detected following foreign body ingestion. Indian J Gastroenterol. 1996;15(3):109–10.

Otjen JP, Iyer RS, Phillips GS, Parisi MT. Usual and unusual causes of pediatric gastric outlet obstruction. Pediatr Radiol. 2012;42(6):728–37.

Ozge A, Safak H, Ebru H, Evrim U, Bilge SE, Leyla O, Kemal KA, Volkan B. First successful preimplantation genetic diagnosis of epidermolysis bullosa with pyloric

atresia: case study of a novel c.4505-4508insACTC mutation. J Assist Reprod Genet. 2012;29(4):347–52.
Parshotam G, Ahmed S, Gollow I. Single or double bubble: sign of trouble! Congenital pyloric atresia: report of two cases and review of literature. J Paediatr Child Health. 2007;43(6):502–3.
Parsons LG, Barling S. Diseases of infancy and childhood. London: Oxford Medical Publication; 1933. p. 739.
Polonkai E, Nagy A, Csízy I, Molnár C, Roszer T, Balla G, Józsa T. Pyloric atresia associated with Dieulafoy lesion and gastric dysmotility in a neonate. J Pediatr Surg. 2011;46(10):E19–23.
Salvestrini C, McGrath JA, Ozoemena L, Husain K, Buhamrah E, Sabery N, Leichtner A, Rufo PA, Perez-Atayde A, Orteu CH, Torrente F, Heuschkel RB, Thomson MA, Murch SH. Desquamative enteropathy and pyloric atresia without skin disease caused by a novel intracellular beta4 integrin mutation. J Pediatr Gastroenterol Nutr. 2008;47(5):585–91.
Scheida N, Wales PW, Krishnamurthy G, Chait PG, Amaral JG. Ectopic drainage of the common bile duct into the lesser curvature of the gastric antrum in a newborn with pyloric atresia, annular pancreas and congenital short bowel syndrome. Pediatr Radiol. 2009;39(1):66–9.
Sencan A, Mir E, Karaca I, Günşar C, Sencan A, Topçu K. Pyloric atresia associated with multiple intestinal atresias and pylorocholedochal fistula. J Pediatr Surg. 2002;37(8):1223–4.
Sharma KK, Ranka P, Goyal P, Dabi DR. Gastric outlet obstruction in children: an overview with report of Jodhpur disease and Sharma's classification. J Pediatr Surg. 2008;43(10):1891–7.
Tiao MM, Ko SF, Hsieh CS, Ng SH, Liang CD, Sheen-Chen SM, Chuang JH, Huang HY. Antral web associated with distal antral hypertrophy and prepyloric stenosis mimicking hypertrophic pyloric stenosis. World J Gastroenterol. 2005;11(4):609–11.
Tomá P, Mengozzi E, Dell'Acqua A, Mattioli G, Pieroni G, Fabrizzi G. Pyloric atresia: report of two cases (one associated with epidermolysis bullosa and one associated with multiple intestinal atresias). Pediatr Radiol. 2002;32(8):552–5.
Usui N, Kamiyama M, Kimura T, Kamata S, Nose K, Fukuzawa M. Prenatal diagnosis of isolated congenital pyloric atresia in a sibling. Pediatr Int. 2013;55 (1):117–9.
Yokoyama S, Utsunomiya H. A case of successful surgical repair for solid segment type pyloric atresia using a novel gastroduodenostomy procedure. J Pediatr Surg. 2012;47(11):2158–60.
Yu DC, Voss SD, Javid PJ, Jennings RW, Weldon CB. In utero diagnosis of congenital pyloric atresia in a single twin using MRI and ultrasound. J Pediatr Surg. 2009;44 (11):e21–4.
Zecca E, Corsello M, Pintus C, Nanni L, Zecca S. Peculiar type 1 congenital pyloric atresia: a case report. Ital J Pediatr. 2010;36:3.