Pyloric atresia and Down's syndrome: prenatal double bubble false sign

M.E. Kuan Arguello, R.M. Martín-Crespo, H.J. Ramírez Velandia, N. Carrera Guermeur, M.C. López Menau[1], P. Maruszeswki, A. Pantoja Bajo[1], J.C. Moreno de Prado[2], R. Luque Mialdea

Pediatric Surgery Department. [1]Neonatology Unit. [2]Fetal Medicine Unit. Gynecology and Obstetrics Department. Toledo Hospital Complex.

ABSTRACT

Objective. Pyloric atresia is a rare malformation, with an incidence of 1:100,000 live newborns. Male to female ratio is 1/1. Typically, it is an isolated malformation, with a good prognosis, but 20-40% of cases present epidermolysis bullosa, and to a lesser extent, multiple intestinal atresias.

We present the case of a pre-term newborn prenatally diagnosed with polyhydramnios, duodenal atresia with "double bubble" sign, and suspected Down's syndrome, who eventually had pyloric atresia.

KEY WORDS: Pyloric atresia; Down's syndrome; Prenatal diagnosis; Double bubble.

ATRESIA PILÓRICA Y SÍNDROME DE DOWN: FALSO SIGNO DE LA DOBLE BURBUJA PRENATAL

RESUMEN

La atresia pilórica es una malformación rara, presenta una incidencia de 1:100.000 recién nacidos vivos y la ratio hombre/mujer es de 1/1. Generalmente es una malformación aislada, con buen pronóstico, pero entre el 20-40% de los casos se asocia a epidermólisis bullosa y en menor frecuencia a otras atresias intestinales múltiples.

Presentamos un caso de recién nacido pretérmino con atresia pilórica con el diagnóstico prenatal de polihidramnios, atresia duodenal con signo de 'doble burbuja' y sospecha de síndrome de Down.

PALABRAS CLAVE: Atresia pilórica; Síndrome de Down; Diagnóstico prenatal; Doble burbuja.

INTRODUCTION

The etiology of pyloric atresia (PA) is unknown. Various embryological theories have been proposed, and according to some authors, it is caused by a disorder in development occurring at intrauterine weeks 5-12[1,2]. It is a rare malformation accounting for 1% of intestinal atresias, with an incidence of 1:100,000[3]. Trisomy 21 is present in 1 out of 1,000 live newborns, which means the probability of both occurring concomitantly is 1:100,000,000[4]. Up until now, only three cases had been described in the literature –ours is the fourth.

The ongoing advances made in prenatal diagnostic techniques allow PA to be diagnosed pre-birth through ultrasonography and MRI. However, if gastric peristalsis is not carefully examined, a false diagnosis of duodenal atresia may be achieved.

CLINICAL CASE

Female neonate born at gestation week (GW) 33+3. Pregnancy had been uneventful but subject to controls. Triple aneuploidy screening in the first three gestation months showed a high risk of trisomy 21 (1:50). However, the parents refused prenatal genetic testing.

At GW 30, severe polyhydramnios was observed, with suspected obstruction of the digestive tract. In addition, serial prenatal ultrasonography at GW 31-32 showed a "double bubble" gastric abdominal image and a 57 x 22 mm gastric image. An adjacent anechoic image potentially suggestive of duodenal atresia was also found. Amniotic fluid index was 36. Fetal Doppler pattern was type III (Fig. 1).

At GW 33 + 3, given that fetal well-being was being compromised, an elective Cesarean section was carried out, with previous fetal maturation. The baby was born with a Down phenotype. She weighed 1,925 g at birth, and somatic maturity was estimated at 33 weeks. She presented widespread hypotonia and restrictive muscular interventricular communication, without hemodynamic repercussion. Karyotype was 47XX + 21.

In the immediate postnatal period, she had respiratory distress treated with CPAP (30% FiO_2). She required endotracheal intubation and surfactant. Light, non-bilious

Corresponding author: Dra. María Esmeralda Kuan Arguello.
E-mail: mari.kuan@gmail.com; mari_kuan@hotmail.com

Date of submission: June 2020 *Date of acceptance:* December 2020

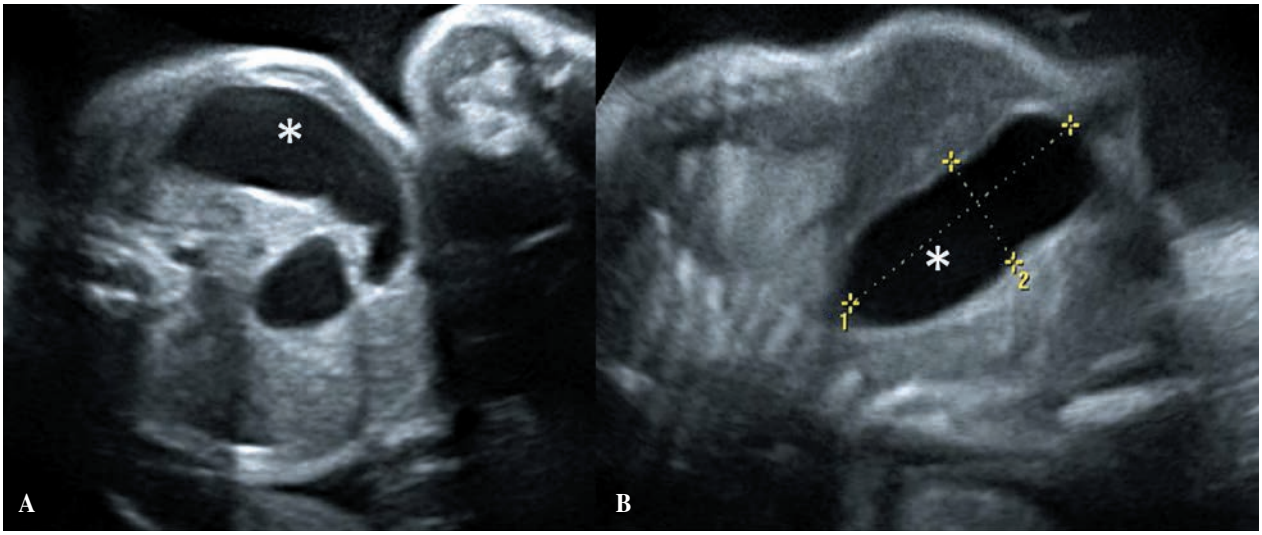

Figure 1. Prenatal ultrasonography at gestation week 32. A) Transverse cut demonstrating a double bubble image. B) Sagittal cut demonstrating a 57 x 22 mm gastric image (*gastric pouch).

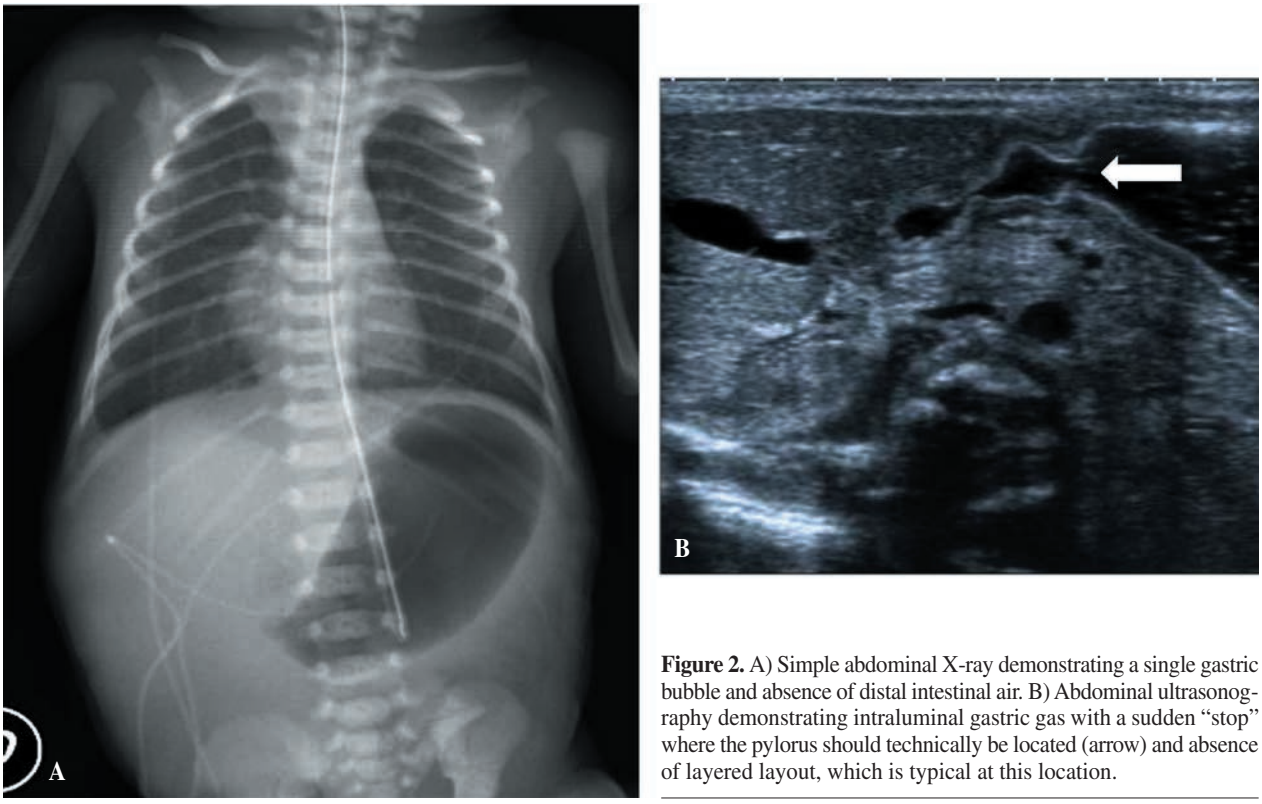

Figure 2. A) Simple abdominal X-ray demonstrating a single gastric bubble and absence of distal intestinal air. B) Abdominal ultrasonography demonstrating intraluminal gastric gas with a sudden "stop" where the pylorus should technically be located (arrow) and absence of layered layout, which is typical at this location.

material came out through the nasogastric tube (NGT). At 12 hours of life, simple abdominal X-ray showed a single gastric bubble and absence of distal intestinal air (Fig. 2A). Abdominal ultrasonography showed gastric intraluminal gas with a sudden "stop" where the pylorus should technically be located (arrow) and absence of layered layout, which is typical at this location (Fig. 2B). 30 cm^3 of saline solution were administered through the NGT, which demonstrated gastric pouch repletion, with the content not being able to progress.

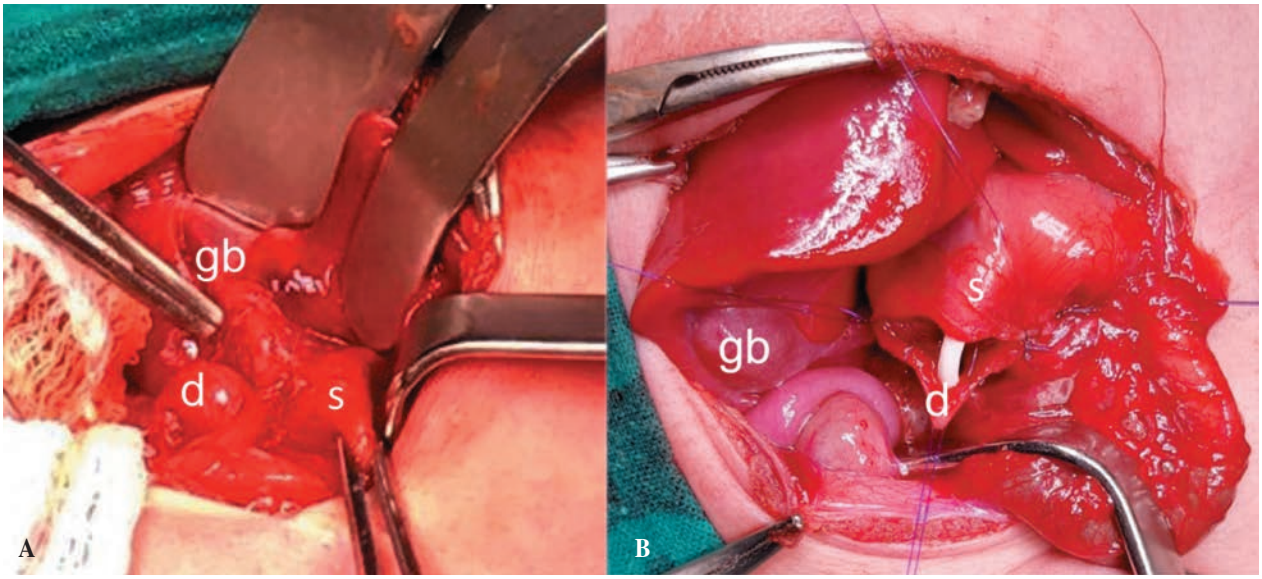

Figure 3. Surgical pictures of supraumbilical transverse laparotomy: A) Large stomach (s) with a round-shaped ending and no continuity with the duodenum (d), type C pyloric atresia (gb: gallbladder). B) Diamond-shaped gastroduodenostomy with anastomotic guiding using a transanastomotic tube.

Diagnosis of pyloric atresia was established. A supraumbilical transverse laparotomy was indicated. It showed a large stomach –in a normal position– and pyloric atresia, with a round-shaped ending, no continuity with the duodenum (type C pyloric atresia), and intestinal malrotation without cecum fixation and with a fixed loop in a normal position (Fig. 3A). A diamond-shaped gastroduodenostomy was carried out, with anastomotic guiding using a transanastomotic tube (Fig. 3B).

In the immediate postoperative period, enteral nutrition through the transanastomotic tube was initiated on day 5, and on day 19, oral nutrition was started. Control digestive transit assessment showed severe gastroesophageal reflux (GER) and a nice gastroduodenal passage. GER medical treatment –both posture and drug-based– was initiated, and subsequent progression was good.

At 9 months of age, GER persists, but the patient has no respiratory symptoms. Oral nutrition and GER medical treatment are maintained. Weight and size gains are normal.

DISCUSSION

PA was first described by Calder in 1749. In 1940, Touroff, Sussman, Meltz et al. reported the first surgical repair[1]. PA may occur as a single malformation (35%), or associated with epidermolysis bullosa (EB) (40%) or other abnormalities (25-55%), such as multiple intestinal atresias, renal and urethral abnormalities, cardiac malformations, aplasia cutis, onychodystrophy, and gallbladder agenesis[5].

The association of PA and EB is known as Carmi's syndrome, which has high morbidity and mortality rates. It has recessive autosomal inheritance, which means parents should receive genetic counselling[6].

PA can also be associated with multiple intestinal atresia – an extremely rare condition which can potentially be inherited. Atresias may involve the whole gastrointestinal tract, from the esophagus to the rectum. In the literature, esophageal atresia is the most commonly reported type of intestinal atresia associated with PA[3].

According to the malformation's anatomical type, PA can be classified into type A –a membrane (57%)–, type B –pyloric tissue replaced by a solid tissue cord (34%)–, and type C –separation between the stomach and the duodenum (9%). Our patient had type C PA[3].

Prenatal diagnosis is achieved through ultrasonography and/or MRI in 50-55% of cases. It is characterized by severe polyhydramnios and progressive gastric pouch dilatation, with a single bubble image, which is characteristic. Some authors have reported esophageal dilatation, and Doppler ultrasonography has revealed gastroesophageal reflux[7]. In some cases, a double bubble sign can be observed in PA patients. It is typical of duodenal atresia, it is caused by gastric pouch and duodenal bulb dilatation secondary to duodenal obstruction, and it is visualized in transverse cuts[8]. In our case, owing to the large size of the stomach and the action of gastric peristalsis, a C-shaped gastric pouch could be observed in the transverse cut, which proved to be a false sign of double bubble image. To avoid this, Yoshizato T et al. recommend uninterrupted ultrasound observation for 60 minutes, since this allows

gastric peristaltic periods to be assessed, and actual double bubble to be distinguished from the presence of a single gastric pouch[9]. Once gastric obstruction is suspected, it is important to determine comorbidities, since PA is frequently associated with epidermolysis bullosa. In addition, during ultrasound exploration, the "snowflake" sign and turbid amniotic fluid –which is caused by the presence of squames, a cutaneous sign characteristic of EB-type cutaneous disorders– should be searched for[10].

In the absence of prenatal diagnosis, pyloric atresia occurs in the form of non-bilious vomit, upper abdominal distension, and hydroelectrolytic disorder. Gastric perforation instances have also been described. Diagnosis is suspected by means of a simple abdominal X-ray, which shows a single air bubble without distal gas. It is complemented with a gastrointestinal transit test or with ultrasonography –as in our case–, which demonstrates an unusual distribution of pyloric layers and absence of gastric content progression. In some type A cases (pyloric membrane), diagnosis has been achieved by means of upper GI endoscopy[11].

Once diagnosis has been confirmed, a decompressive nasogastric tube should be placed, with an adequate repair of the hydroelectrolytic disorder[2]. Treatment is always surgical, with the surgical technique varying according to the anatomical type. In type A, membrane resection and Heineke-Mikulicz or Finney pyloroplasty are carried out. In type B, Dessanti et al. described a reconstruction technique for the pyloric sphincter, but gastroduodenostomy is the gold standard procedure[12]. In type C, gastroduodenostomy is indicated. During surgery, other intestinal atresias or associated malformations should be ruled out. Placing a transanastomotic tube is useful for early enteral nutrition initiation[13].

Consistent with the literature, our patient had the usual postoperative complications following PA surgery – delayed gastric emptying and GER, which responded to medical treatment. No cases requiring surgical GER treatment have been reported, but long-term follow-up is recommended to evaluate the effects of biliary reflux[14].

PA has a mortality rate of approximately 50% as a result of the association with other conditions. In the absence of EB and other associated abnormalities, PA can be surgically repaired and has a good prognosis[14].

CONCLUSION

Even though it has a very low prevalence, well below that of duodenal obstruction, PA should be considered as part of the differential diagnosis in the presence of a double bubble sign. At prenatal ultrasonography, the peristaltic wave should be carefully observed in the whole gastric pouch, since it may show a false double bubble image. Early diagnosis and surgery, as well as neonatal intensive care improvements, have allowed for a better prognosis in PA patients without further pathologies.

REFERENCES

1. Ilce Z, Erdogan E, Kara C, Celayir S, Sarimurat N, Senyüz OF. Pyloric Atresia: 15-year review from a single institution. J Pediatr Surg. 2003; 38(11): 1581-4.
2. Andriessen MJ, Matthyssens LE, Heij HA. Pyloric atresia. J Pediatr Surg. 2010; 45: 2470-2.
3. Al-Salem AH, Abdulla MR, Kothari MR, Naga MI. Congenital pyloric atresia, presentation management, and outcome: A report of 20 cases. J Pediatr Surg. 2014; 49: 1078-82.
4. Patel SB, Milstein JM, Schwartz MZ, Wood BP. Radiological cases of the month: Congenital Pyloric Atresia in Down Syndrome. Am J Dis Child. 1993; 147(3): 307-8.
5. Kansra M, Raman VS, Kishore K, Khanna S, Puri B, Sharma A. Congenital pyloric atresia- nine new cases: Single center experience of the long term follow-up and the lessons learnt over a decade. J Pediatr Surg. 2018; 53: 2112-6.
6. Gupta R, Soni V, Mathur P, Goyal RB. Congenital pyloric atresia and associated anomalies: A case series. J Neonatal Surg. 2013; 4: 40.
7. Rizzo G, Capponi A, Arduini D, Romanini C. Prenatal diagnosis of gastroesophageal reflux by color and pulsed Doppler ultrasonography in a case of congenital pyloric atresia. Ultrasound Obstet Gynecol. 1995; 6: 290-2.
8. Malone FD, Crombleholme TM, Nores JA, Athanassiou A, D'Alton ME. Pitfalls of the 'Double Bubble' Sign: A Case of Congenital Duodenal Dublication. Fetal Diagn Ther. 1997; 12: 298-300.
9. Yoshizato T, Satoh S, Taguchi T, Suita S, Nakano H. Intermittent 'Double Bubble' Sign in a Case of Congenital Pyloric Atresia. Fetal Diagn Ther. 2002; 17: 334-8.
10. Lepinard C, Descamps P, Meneguzzi G, Blanchet-Bardon C, Germain DP, Larget-Piet L. Prenatal diagnosis of pyloric atresia-junctional epidermolysis bullosa syndrome in a fetus not known to be at risk. Prenat Diagn. 2000; 20: 70-5.
11. Okoye BO, Parikh DH, Lander AD. Pyloric Atresia: Five new cases, a new association, and a review of the lierature with guidelines. J Pediatr Surg. 2000; 35: 1242-5.
12. Dessanti A, Benedetto VD, Iannucceli M, Balata A, Rocca PC, Benedetto AD. Pyloric atresia: a new operation to reconstruct the pyloric sphincter. J Pediatr Surg. 2004; 39(3): 297-301.
13. Heinen FL. Atresia de píloro. Comunicación de 3 casos. Arch Argent Pediatr. 2014; 112(5): e227-e230.
14. Schutzman LM, Walsh M, Draus JM Jr. Pyloric atresia: Comorbidity determines Outcome. J Ped Surg Case Reports. 2014; 2: 192-5.