Journal of Pediatric Surgery 49 (2014) 1078–1082

Contents lists available at ScienceDirect

Journal of Pediatric Surgery

journal homepage: www.elsevier.com/locate/jpedsurg

Congenital pyloric atresia, presentation, management, and outcome: A report of 20 cases

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ARTICLE INFO

Article history:
Received 1 July 2013
Received in revised form 7 October 2013
Accepted 8 October 2013

Key words:
Pyloric atresia
Epidermolysis bullosa
Aplasia cutis congenita
Hereditary multiple intestinal atresia

ABSTRACT

Background: Congenital pyloric atresia (CPA) is a very rare anomaly. It is usually seen as an isolated condition with excellent prognosis. Few cases are familial. These are usually associated with other hereditary conditions and have a poor prognosis. This is a review of our experience with 20 patients with CPA outlining aspects of diagnosis, associated anomalies and management.

Patients and methods: This is a retrospective analysis of 20 cases seen over a 22 year period (December 1990 to December 2012). Their records reviewed for: age, sex, presentation, prenatal history, associated anomalies, investigations, treatment, operative findings and the outcome.

Results: 20 cases (9 Males, 11 Females) were treated. 7 patients were full term and the remaining 13 were prematures. Their mean birth weight was 2.1 kg (1.1 kg to 3.9 kg). Polyhydramnios was seen in 13 patients (65%). Two were brothers and four were members of the same family. Isolated CPA was seen in 7 (35%); 13 had an associated conditions: epidermolysis bullosa (EB) in 8 (40%) and multiple intestinal atresias (MIA) in 5 (25%). Three patients had associated esophageal atresia. All were operated on except two who died early due to unrelenting sepsis. The variety of pyloric atresias encountered were as follows: pyloric diaphragm in 13 including double diaphragms in 2, pyloric atresia with a gap in 4 and pyloric atresia without gap in 3. Ten died postoperatively giving an overall survival of 40%.

Conclusions: CPA is a very rare condition. Isolated CPA carries a good prognosis. Association of CPA with other familial and congenital anomalies like EB and MIA carries a poor prognosis.

Congenital pyloric atresia (CPA) is a very rare condition that was first described by Calder in 1749 [1–3]. It accounts for less than 1% of all upper gastrointestinal atresias with a reported incidence of 1:100,000 live births [1–3]. CPA may occur as an isolated lesion or in association with other congenital or hereditary anomalies. The association of Epidermolysis bullosa (EB) with CPA is common and has been reported in several publications [4–8]. The diagnosis and management of isolated CPA is simple but when CPA is associated with other anomalies, diagnosis may be either missed or delayed and the management is complicated. The presence of associated anomalies is a contributing factor for the reported high mortality [9]. This is a review of our experience in 20 patients with CPA outlining aspects of diagnosis, associated anomalies and management. The influence of associated anomalies on outcome is also discussed.

1. Materials and methods

This is a retrospective analysis of 20 cases seen over 22 year period (December 1990–December 2012) at our institute. The records of the patients with the diagnosis of CPA were reviewed and following parameters were recorded: Age, sex, presentation, prenatal history, associated anomalies, investigations, treatment, operative findings and the outcome.

2. Results

Twenty cases (9 males, 11 females) with CPA were treated. Seven patients were full term and the remaining 13 were premature babies. Their mean birth weight was 2.1 kg (1.1 kg to 3.9 kg). Polyhydramnios was seen in 13 patients (65%). Two of our patients were brothers, one of them had isolated CPA while the other had CPA associated with duodenal atresia, jejunal atresia and a duplication cyst [12]. Four were members of the same family, all had associated EB. Isolated CPA was seen in 7 (33.3%) patients. Eight (40%) had CPA associated with EB. One of these also had aplasia cutis congenita affecting multiple areas of varying size on left lower leg, dorsum of left foot, left knee, left ear, right ear, neck, bridge of nose, right ankle and back of the right knee. Other associated anomalies seen were Down's syndrome in one, congenital heart disease in one and cleft palate in another (Tables 1, 2 and 3).

In five patients CPA was associated with multiple intestinal atresias (MIA). One of them had atresia at duodeno-jejunal junction,

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http://dx.doi.org/10.1016/j.jpedsurg.2013.10.009
0022-3468/

Table 1
Clinical features of patients with congenital pyloric atresia associated with epidermolysis bullosa.

No.	Age	Sex	BW	GA	Associated Anomalies	Type of atresia	Treatment	Outcome
1.	6 days	F	3.2 kg	Full term	EB	Double pyloric diaphragm	Excision and Pyloroplasty	Survived
2. (F)	2 days	F	2.45 kg	Preterm	EB	Pyloric Diaphragm	Excision and pyloroplasty	Died at 2 months from sepsis
3. (F)	2 days	M	2.1 kg	Preterm	EB	Pyloric Diaphragm	Excision and pyloroplasty	Died at 1 months from sepsis
4. (F)	2 days	M	2.5	Preterm	EB	Pyloric Diaphragm	Excision and pyloroplasty	Died at 3 month from sepsis
5. (F)	3 days	F	1.8 kg	Preterm	EB	Pyloric Diaphragm	Excision and pyloroplasty	Died at 9 months from sepsis
6.	2 days	M	1.5 kg	Preterm	EB ACC	Pyloric atresia with a gap	Gastroduodenostomy	Died at 27 days from sepsis
7.	1 day	M	1.75 kg	Preterm	EB EA + TOF Cleft palate	Pylroic atresia	Gastrostomy Gastrojejunostomy	Died at 9 days from sepsis
8.	2 days	F	1.85 kg	Preterm	EB	No surgery	None	Died at 16 days from sepsis

E.B = epidermolysis bullossa, A.C.C = aplasia cutis congenital, EA = esophageal atresia, TOF = tracheoesophageal fistula. (F) = members of the same family.

and malrotation with ileal perforation [10]. Closed loop obstruction of the duodenum resulted in duodenal perforation in this patient.

Three patients had CPA associated with esophageal atresia. Two of these patients had esophageal atresia with tracheoesophageal fistula and an anorectal malformation. In both patients the diagnosis of CPA was considered preoperatively due to presence of a single air bubble on plain abdominal x-ray associated with esophageal atresia and tracheoesophageal fistula (Fig. 1). The third patient had pure esophageal atresia, CPA and EB. In this patient, CPA was diagnosed intraoperatively at the time of gastrostomy. The presence of EB in this patient hinted at the possibility of associated CPA.

All patients underwent surgical correction except two who died preoperatively. Both patients suffered from multiple episodes of sepsis due to variety of organisms. The types of CPA encountered were: pyloric diaphragms in eleven patients (two of which were double diaphragms), pyloric atresia with a gap in four patients and pyloric atresia without gap in three patients (Fig. 2). Excision of diaphragm and Heineke-Mikulicz pyloroplasty was done in patients with pyloric diaphragms. Five patients underwent gastro-duodenostomy and duodeno-jejunostomy was done in one patient with associated distal duodenal atresia along with excision of duplication cyst. In addition to gastro-duodenostomy and duodeno-jejunostomy, the patient with duodenal and ileal perforation also underwent duodenoplasty as the duodenum was markedly dilated. The area of ileal perforation was resected and end to end anastomosis was done after correction of the associated malrotation. Regarding the three patients with associated esophageal atresia, one with pure atresia had gastrostomy and gastro-jejunostomy, one underwent ligation of the fistula, gastrostomy and colostomy for associated anorectal malformation and one patient had ligation of fistula, end to end esophageal anastomosis, gastro-duodenostomy and anal transposition for associated anterior ectopic anus. The last patient did well postoperatively and is currently being followed. All the operated cases survived in the early postoperative period but subsequently 12 patients died resulting in overall survival of 40%. Sepsis was the main cause of death.

3. Discussion

Congenital anomalies of the gastrointestinal tract are relatively common, but CPA is extremely rare contributing less than 1% of all upper gastrointestinal atresias [1–3]. Commonly, it occurs as an isolated lesion which presents with upper abdominal distension and non-bilious vomiting. The diagnosis of CPA is suggested by an abdominal x-ray which classically shows a large single gastric air-bubble and a gasless non-distended abdomen. A perforated pyloric web with scanty gas distally in the gastrointestinal tract has also been reported [11]. Such a presentation may need an upper contrast study or endoscopy to confirm the diagnosis. An antenatal ultrasound in the second trimester may suggest CPA in presence of polyhydramnios and a dilated stomach [6,12,13]. Contrast studies can be used to confirm the diagnosis but are not usually necessary (Fig. 3). Neonatal gastric perforation is a rare and unusual presentation of CPA.

Although seen usually as an isolated anomaly, CPA can also occurs in association with other genetic disorders like EB and aplasia cutis congenita (ACC). Multiple other gastrointestinal tract atresias are also seen with some frequency in these patients [4,9–11,14–18]. CPA patients with these associations are reported to have high mortality. Moore in 1989 reviewed the literature and reported 125 cases of CPA [1]. Three cases had associated esophageal atresia and eighteen had EB. In this report we describe 20 newborns with CPA. Isolated CPA occurred in seven (35%) patients, eight patients (40%) had associated EB and 5 patients (25%) had associated other gastrointestinal atresias. This divides CPA into three clinical types: Isolated CPA, CPA associated

Table 2
Clinical features of patients with congenital pyloric atresia associated with other intestinal atresias.

No.	Age	Sex	BW	GA	Associated Anomalies	Type of atresia	Treatment	Outcome
1. (B)	1 day	M	1.38 kg	Full term	Jejunal atresia Duplication cyst	Pyloric atresia with gap	Gastroduodenostomy + dudenojejunostomy + excision of duplication cyst	Died at 3 days
2.	1 day	M	2.6 kg	Full term	Duodenal perforation Ileal perforation Jejunal atresia Malrotation	Pyloric atresia with a gap	Gastroduodenostomy Duodenoplasty Duodenojejunostomy Ileal resection Ileoileal anastomosis	Died at 31 day
3.	1 day	F	2.4 kg	Full term	Down's syndrome CHD	No surgery	None	Died at 38 days
4.	6 days	M	1.1 kg	Preterm	EA + TOF ARM Single Umbilical art. High arched palate	Pylroic diaphragm	Ligation of TOF, gastrostomy and colostomy	Died at 6 days
5.	2 days	F	2.4 kg	Preterm	EA + TOF ARM	Pyloric atresia with gap	Ligation of fistula, esophageal end to end anastomosis, gastroduodenostomy and anal transposition	Survived

EA = esophageal atresia, TOF = tracheoesophageal atresia, ARM = anorectal malformation, CHD = congenital heart disease. B = brothers.