Case Report

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Pyloric atresia–Three cases and review of literature

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ABSTRACT

Pyloric atresia (PA) is a rare congenital anomaly that constitutes approximately 1% of all intestinal atresias, and its incidence is approximately 1 in 100,000 live births. PA may occur as an isolated condition or associated with other abnormalities, the most common being Junctional epidermolysis bullosa (JEB). Evidence suggests that PA-EB (Pyloric Atresia - Epidermolysis Bullosa) Syndrome is a distinct entity. In this report, we present three cases of pyloric atresia, one of which was associated with Junctional epidermolysis bullosa. The literature on the subject is also reviewed.

Key words: Gastroduodenostomy, PAEB syndrome, pyloric atresia

INTRODUCTION

Pyloric atresia (PA) is a rare malformation which is estimated to be responsible for less than 1% of gastro-intestinal atresia's with an incidence of 1 in 100,000 live births.[1] Epidermolysis bullosa (EB) consists of a group of hereditary skin disorders, where the primary cause is formation of blisters following minor trauma. The recessive form of EB has a 1 in 300,000 incidence. EB is divided into 3 main types according to the histopathologic location of the bullae. Epidermolysis bullosa simplex (EBS) has been reported frequently in association with. PA.[2] Evidence suggests that PA-EB association is a distinct entity and is now referred to as the PAEB syndrome.[3] We hereby present three cases of PA one of which was associated with EB.

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CASE REPORTS

Case 1

A three day old girl, born by Caesarean section at 32 weeks of gestational age, presented with copious, non-bilious vomiting since birth with mild epigastric dilation. Antenatal sonography was normal and there was no polyhydramnios. Physical examination showed multiple bullous eruptions over all extremities [Figure 1]. An abdominal X-ray showed distended stomach with no distal gas. Laparotomy revealed PA type 2 and gastroduodenostomy was performed [Figure 2]. Patient



Figure 1: Multiple skin eruptions in epidermolysis bullosa

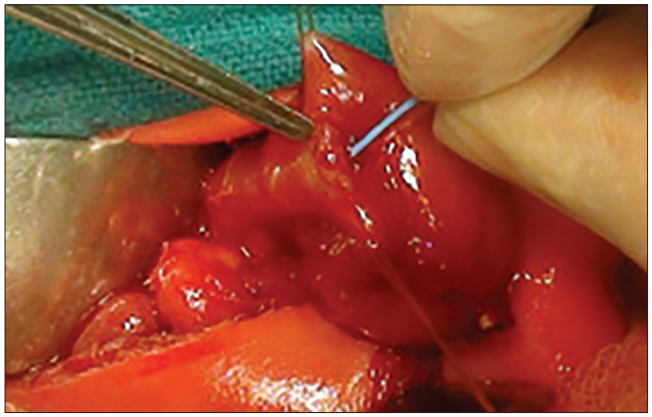


Figure 2: Operative photograph showing pyloric atresia type 2

started feeds on post-operative day five, which was tolerated well and hence increased gradually to full feeds. However, she developed extensive skin infection with septicaemia and eventually succumbed on post-operative day 12.

Case 2

A 14 m girl came with complaints of recurrent non-bilious vomiting since birth. Antenatal ultrasound (US) was normal. Upper abdomen was grossly distended with visible gastric peristalsis. US were suggestive of grossly distended stomach with no obvious hypertrophy of pylorus. Abdominal plain X-ray showed single gas bubble. [Figure 3] Gastroscopy revealed tight pyloric stenosis, scope could not be negotiated. Upper GI contrast study revealed dilation of stomach with no contrast beyond the pylorus. [Figure 4] At laparotomy pyloric end was firm and whitish with mucosal diaphragm. The pyloric membrane was excised and Heineke- Mikulicz pyloroplasty performed. Patient recovered well and is doing well on follow-up.



Figure 3: X-Ray abdomen erect showing single bubble



Figure 4: Upper GI Contrast study showing pyloric obstruction

Case 3

A 10 day old boy, born by emergency caesarean section at 34 weeks gestation, presented with non-projectile and non-bilious vomiting since birth. Physical examination revealed shrill cry with signs of dehydration and sepsis. Routine haematological examinations showed increased C-reactive protein and total counts. Upper abdomen was mildly distended. X-ray abdomen showed a single gastric bubble with no air in the small and large intestine. At laparotomy there was a moderately distended stomach and a narrowed duodenal tract. In view of frank sepsis and poor general condition of the child, gastro-jejunostomy was done. The child was on ventilator for 5 days and was gradually weaned off. He was treated with total parenteral nutrition for 10 days. Oral feeding was started on postoperative day 7 without problems. He is well now at 12m of life.

DISCUSSION

The earliest description of PA is credited to Caleder in 1733,[4] and Swinbure and Kohler first described the association of PA and EB in 1968.[5] Ninety-one cases of this rare syndrome have been reported in the literature so far.[3] Anatomically, PA is divided into 3 types pyloric membrane or web type I (57%), pyloric canal replaced by a solid cord of tissue type II (34%), PA with a gap or gap aplasia type III (9%).[6] Carmi suggested the pathophysiology of the disorder; therefore, it is also referred to as 'Carmi syndrome'.[7] Both PA and EB are very rare entities. The combination of these pathologies appears more than coincidental and is not the result of chance association.[8] There are many theories regarding the cause of PA in patients with PA-EB. Chang *et al*. proposed that PA occurs secondary to an intrauterine complication of EB where sloughing pyloric mucosa leads to fibrosis and obstruction of the pyloric canal.[9]

The typical presentation of PA is non-bilious vomiting soon after birth without abdominal distention. The diagnosis is made easily on plain abdominal X-ray, which shows a single gas bubble representing the distended stomach with no distal gas. Most of these patients are premature as were our two neonates. Diagnosis of EB is based on clinical and laboratory criteria. Major ultrasound sign of PA is gastric dilatation. Antenatal ultrasound may show polyhydramnios and dilated stomach.[10] In 1990 Meizner and Carine described the snow flake sign, an echogenic appearance of the amniotic fluid during the second trimester of pregnancy associated with EB.[11] In the literature most of the prenatal diagnoses have been reported in cases with a positive family history of PA-EB. The association of PA

with EB is well documented and a plain abdominal X-ray should be performed in all babies with EB to exclude PA. The poor prognosis associated with this syndrome has led some attempt to diagnose this condition prenatally when there is a positive family history.

EB is an autosomal recessive gene affecting the integrity of the basement membrane and hemidesmosomes and the control of the normal process of fibrosis occurring during the course of wound healing. The lesions constitute of blisters, crusted erosions and scar formation. There are 3 types of EB based on histopathology of which Junctional EB (JEB) is mostly associated with PA. Defects in ITGA6 or ITGB4 genes have been identified in JEB.[12]

Treatment of PA is surgical. It depends on the type of atresia. In type 1 PA, membrane is excised and Heineke-Mikulicz pyloroplasty performed.[13] For type 2 atresia, excision of atretic segment and gastroduodenostomy is done.[13] For type 3, gastroduodenostomy is performed. Intraoperatively, it is important to locate the site of obstruction especially in those with a diaphragm, and to obviate missing a windsock diaphragm, a catheter should be passed distally *via* a small gastrostomy. This is also of importance in case there is more than one diaphragm.[14] Type 1 atresia predominates in non-EB cases, and type 2 cases are mostly associated with EB. Our first and third patients had type 2 PA and second patient had type 1 PA. Though the surgical management for PA is straightforward, septicaemia, electrolyte imbalance, protein loss and failure to thrive complicates the severe exudative skin lesions, often leading to death in PA-EB. As in our *case 1*, most of the PA/EB infants die within the first few weeks or months of life despite successful surgical correction of PA. Smad *et al*. reported 3 cases of PA-EB. One of them died before operation and one of the two operated children survived. They concluded that although the prognosis is poor in this condition, surgery should not be withheld on assumption of high mortality.[3]

All neonates with pyloric atresia should be screened for other anomalies, in particular, oesophageal atresia and abnormalities of the heart, face, skeleton and genitourinary tract as a part of the hereditary multiple intestinal atresias (HMIA).[6,14] A variety of urinary tract diseases like ureterovesical junction obstruction leading to the early development of hydronephrosis should be suspected. So ultrasonography and Cystoscopy are recommended in all survivors with PA-EB association. Wallerstein *et al*. reported 2 cases of epidermolysis bullosa-pyloric atresia-obstructive uropathy (EB-PA-OU).[15] The syndrome of EBPA- OU is a rare genetic entity of variable severity manifested by skin findings of multiple bullae and infrequently, aplasia cutis congenita, the gastrointestinal finding of pyloric or other atresias, and progressive obstructive uropathy. None of our three patients had urologic problems.

Pyloric atresia (PA)/EB is a highly lethal combination and death is almost a universal result. Hayashi *et al*.[8] reported four patients who had a survival between 17 m to 16 years, but they might have had alleles with lesser likelihood of lethality. Therefore, non-operative management of PA is inappropriate in the stable neonate with EB.

Conclusion - Pyloric Atresia is a rare condition. Pyloric atresia with Epidermolysis bullosa has a high mortality. Every attempt should be made to diagnose this condition prenatally, when there is a positive family history. Also, plain abdominal X-ray should be performed in all babies with Epidermolysis bullosa to exclude Pyloric Atresia. However, surgical treatment should not be withheld on the presumption of a poor outcome.

SUMMARY

A series of three cases of pyloric atresia is reported, one of which is associated with EB Syndrome. Case 1 – A three day old girl presented with non-bilious vomiting with multiple skin eruptions all over all extremities. X-ray showed distended stomach with no distal gas. Laparotomy revealed PA type 2 and gastroduodenostomy was performed. Child succumbed due to septicaemia. Case 2 – A 14 m girl presented with non-bilious vomiting and distended upper abdomen. USG and X-ray were suggestive of distended stomach. Gastroscopy revealed tight pyloric stenosis. At laparotomy pyloric membrane was excised and Heineke- Mikulicz pyloroplasty performed. Patient is doing well on follow-up. Case 3 – A 10 day old boy presented with non-bilious vomiting since birth. Child had signs of dehydration and sepsis. X-ray abdomen showed distended stomach. At laparotomy there was a narrowed duodenal tract. In view of poor general condition of the child, gastrojejunostomy was done. Child did well on follow-up.

Pyloric atresia must be considered in the differential diagnosis of any high intestinal obstruction in the newborn with bile-free emesis. Plain X-ray in EB syndrome may help to exclude PA. Surgical treatment should not be withheld on the presumption of a poor outcome.

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