

# Pyloric atresia epidermolysis bullosa aplasia cutis syndrome: a case report and literature review

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The association between epidermolysis bullosa and congenital pyloric atresia is rare, but is a known distinct clinical entity with autosomal recessive inheritance. We report on a case of a baby girl born by cesarean section at 37 weeks' gestation to a G7 P7 mother and weighed 2.84 kg. At birth, there was denuded skin over the right leg from the knee joint up to the middle of the right foot. Abdominal radiograph was thus performed and indicated absent gas in the small bowel and single bubble appearance of the stomach. Pyloric atresia was surgically corrected. Electron microscopy of skin biopsies showed junctional epidermolysis bullosa. There was typical nail dystrophy and no corneal lesions; ultrasonography for the renal system was normal. At 4 months of age, the baby required gastrostomy and fundoplication for severe failure to thrive. Six weeks after her second surgical procedure,

the patient presented to the emergency room dead; the parents provided a typical scenario of sudden infant death syndrome at home. Examination indicated no signs of child abuse; the parents refused an autopsy exam for the child. *Ann Pediatr Surg* 9:84–86 © 2013 Annals of Pediatric Surgery.

*Annals of Pediatric Surgery* 2013, 9:84–86

**Keywords:** aplasia cutis syndrome, epidermolysis bullosa, pyloric atresia

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Received 4 December 2012 accepted 20 February 2013

## Introduction

Epidermolysis bullosa with pyloric atresia (EB-PA) is characterized by fragility of the skin and mucous membranes, manifest by blistering with little or no trauma; congenital pyloric atresia (CPA); and ureteral and renal anomalies. The course of EB-PA is usually severe and often lethal in the neonatal period. Although most affected children succumb as neonates, those who survive may have severe blistering with formation of granulation tissue on the skin around the mouth, nose, fingers, and toes, and internally around the trachea. However, some affected individuals have little or no blistering later in life. Additional features shared by EB-PA and the other major forms of EB include congenital localized absence of skin (aplasia cutis congenita), milia, nail dystrophy, scarring alopecia, hypotrichosis, and contractures [1]. Because of the rarity of this syndrome, we report a case of Pyloric Atresia Epidermolysis Bullosa Aplasia Cutis Syndrome that was managed at our institution.

Parents' consent was obtained for publishing all the required data for the case.

## Case report

A baby girl was born by cesarean section at 37 weeks' gestation to a G7 P7 mother; the birth weight of the baby was 2.84 kg. Apgar scores were 8 and 9 at 1 and 5 min, respectively. Pregnancy was uneventful, with no history of polyhydramnios.

The parents are second-degree relatives, with no family history of similar disease.

Physical examination at birth indicated stable vital signs and denuded skin over the right leg from the knee joint up to the middle of the right foot; the ankle area was affected circumferentially (Fig. 1).

The baby was shifted to the maternity ward after birth; she passed good amount of meconium in the first 24 h. She started breast feeding, but vomited light yellowish vomitus that was progressive.

Abdominal radiograph was thus performed and indicated absent gas in the small bowel and single bubble appearance of the stomach (Fig. 2). Upper contrast study indicated a diagnosis of PA (Fig. 3).

A provisional diagnosis was made for Pyloric Atresia Epidermolysis Bullosa Aplasia Cutis Syndrome on the basis of the previously mentioned clinical findings.

The case was posted for surgical repair of PA and skin biopsy was collected as well. Surgical findings indicated type 3 PA, for which gastroduodenostomy was performed. The postoperative period was uneventful. Feeding was started on the fourth postoperative day, which progressed to full feed through a nasogastric tube because of the weak sucking reflex of the baby. Electron microscopy of skin biopsies showed junctional epidermolysis bullosa (JEB) (the diagnosis was reviewed by our colleagues in Hannover, Germany).

There was typical nail dystrophy and no corneal lesions; ultrasonography for the renal system was normal.

The baby was discharged home 68 days after delivery with a fair general condition, but still weak sucking reflexes and poor weight gain as well. The right denuded skin started to improve with frequent blistering and skin peeling in other body parts that was managed with epidermal growth factor and Mebo ointment (Gulf Pharmaceuticals Industries, Ras Al khaima, UAE) with local wound dressing.

She was started on omeprazole and domperidone for gastroesophageal reflux disease for 1 month, but despite

Fig. 1



One-week-old baby. Aplasia cutis congenita on the right leg, with healed epidermolysis bullosa on the face.

Fig. 2



Radiograph of plain erect abdomen at day 2 of birth, showing a single gastric air bubble.

Fig. 3



Upper contrast swallow at day 2 of birth; typical radiological signs of gastric outlet obstruction.

this, she developed recurrent chest infection and failure to thrive, with severe ulcerations in the orolaryngopharynx as well as trachea that required frequent hospital admission with IV antibiotics.

At 4 months of age, the decision was taken to operate upon her for gastrostomy and fundoplication as well; intraoperatively, there were severe adhesions in the peritoneal cavity that were mostly because of her original disease.

The baby started to gain weight and there was no recurrent chest infection, but she still had poor sucking reflexes, with some improvement in the orolaryngeal ulcerations. She was receiving the daily calories through the gastrostomy tube with high-calorie milk.

There was no element of malabsorption on stool examination.

Six weeks after her second surgical procedure, the patient presented to the emergency room dead; the parents provided a typical scenario of sudden infant death syndrome at home.

Examination indicated no signs of child abuse; the parents refused to an autopsy exam of the child.

**Discussion**

The association between EB and CPA is rare, but it is a known distinct clinical entity with autosomal recessive inheritance. The outcome of such an association is universally fatal [2].

Dang *et al.* [3], in their recent elegant study published in 2008, reported three cases and reviewed 46 cases of JEB-PA published in the literature.

In addition to PA ( $n = 49$ ), other commonly reported complications in these patients included nail dystrophy ( $n = 7$ ), enamel hypoplasia ( $n = 4$ ), aplasia cutis congenita

or congenital localized absence of skin ( $n = 6$ ), eye involvement ( $n = 4$ ), ear or nose hypoplasia or atrophy ( $n = 4$ ), urinary tract involvement ( $n = 8$ ), and respiratory involvement ( $n = 5$ ). Many patients die of this disease owing to the extensive denudation of skin, resultant loss of barrier function, fluid and electrolyte problems, and sepsis. Difficulty with oral feeding and diarrhea seem to be common features in many of these cases.

Dang *et al.* [3] summarized a total of 49 cases with JEB-PA, 21 being classified as nonlethal cases and 28 as lethal.

Our case is considered a lethal type of JEB-PA; no studies were carried out to detect any mutations. It was associated with aplasia cutis congenital, nail dystrophy, and respiratory involvement. There was failure to thrive (even after fundoplication and gastrostomy) as well as difficulty in oral feeding, which is a common feature for those cases, attributable to multifactorial reasons. The absence of consanguinity between parents (Syrian nationality) in our case is an uncommon presentation.

From 2008 to 2012, the Pubmed search engine was searched for Pyloric Atresia Epidermolysis Bullosa Aplasia Cutis Syndrome; there were five publications; one of them was a book chapter and four were case reports reporting four cases of JEB-PA [1,4–7].

To our knowledge, to date, a total of 53 cases have been reported in the literature; therefore, our study represents the 54th case.

### Acknowledgements

The authors acknowledge Professor Dr Med. Benno Ure, Direktor des Zentrums Kinderchirurgie Hannover, Dr

B. Ludwikowski, MD, Associate Professor of Pediatric Surgery, Chief of Pediatric Surgery and Pediatric Urology, Auf der Bult, Kinder-u. Jugendkrankenhaus, Hannover, Germany, as well as Dr Anja Diem and Dr Martin Laimer, Salzburg, Austria, for their valuable contribution in histological analysis of the skin biopsy.

### Conflicts of interest

There are no conflicts of interest.

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