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Congenital Pyloric Atresia Associated With a Self-Improving Epidermolysis Bullosa Phenotype

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Abstract

Congenital pyloric atresia (CPA) coexisting with epidermolysis bullosa (EB) represents a rare congenital disorder characterized by complete gastric outlet obstruction and mucocutaneous fragility. Historically, this syndromic association has carried a guarded prognosis, with high neonatal mortality rates secondary to recurrent septicemia, extensive cutaneous denudation, profound fluid and electrolyte losses, and multiorgan failure. We report an unusual presentation in a five-day-old male neonate admitted with persistent non-bilious emesis, lethargy, fever, moderate dehydration, hemoconcentration, and prerenal azotemia. Preoperative abdominal radiography demonstrated a classic single dilated gastric air bubble with complete absence of distal bowel gas, while ultrasonography identified marked gastric distension and an occlusive intraluminal pyloric mucosal diaphragm without muscular hypertrophy, establishing a diagnosis of CPA and excluding infantile hypertrophic pyloric stenosis (IHPS). On day seven of life, the neonate underwent an exploratory laparotomy comprising excision of the complete pyloric mucosal diaphragm and a two-layer Heineke-Mikulicz pyloroplasty. On the eighth postoperative day (day 15 of life), the neonate spontaneously developed multiple tense, flaccid, fluid-filled bullae and superficial erosions at friction-prone acral sites on the left upper and right lower extremities. Intravenous fluid or medication extravasation was carefully excluded, and a clinical diagnosis of EB was established in consultation with the dermatology team. The cutaneous lesions regressed spontaneously over the next two weeks under atraumatic wound care. In view of the rapid resolution suggestive of a self-improving dystrophic epidermolysis bullosa (DEB) phenotype, a skin biopsy for immunofluorescence mapping and a multigene panel were advised; however, both were declined by the parents due to financial constraints, leaving the specific EB subtype as a presumptive clinical diagnosis. Longitudinal follow-up at two years of age confirmed complete cessation of blistering, with only residual postinflammatory hypopigmented patches at previously affected sites, absence of dystrophic scarring or milia, thriving physical growth, and intermittent episodes of ocular pruritus. This case illustrates a rare clinical presentation of type 1 CPA associated with a benign, self-improving blistering phenotype clinically consistent with self-improving DEB.

Categories: Pediatric Surgery
Keywords: dystrophic, epidermolysis bullosa, newborn, pyloric atresia, self improving

Introduction

Congenital pyloric atresia (CPA) is an exceedingly uncommon alimentary tract malformation, representing less than 1% of all congenital gastrointestinal atresias, with an estimated prevalence of one in 100,000 live births [1,2]. When CPA coexists with inherited junctional epidermolysis bullosa (JEB), an association historically designated as Carmi syndrome, it presents an acute clinical and surgical dilemma [3-5]. The disorder has classically been associated with catastrophic neonatal mortality, commonly exceeding 50-70%, predominantly precipitated by overwhelming cutaneous sepsis, profound electrolyte disturbances, and severe malnutrition [5-7].

CPA is classified into three distinct subtypes, namely a type 1 lesion, which is an intraluminal mucosal web or diaphragm across the pyloric canal; a type 2 lesion, which is a fibrous cord replacing the pyloric canal without an anatomical gap; and finally a type 3 lesion, which is a complete segmental gap between the stomach and duodenum [1,2,5]. The pathogenesis of pyloric occlusion in epidermolysis bullosa (EB) is postulated to result from intrauterine sloughing of the mucous membrane with subsequent healing by fibrous tissue leading to progressive fibrosis of the pyloric channel and obliteration of the pyloric lumen [3,4]. While the vast majority of syndromic cases involve JEB linked to ITGB4 or ITGA6 gene alterations or epidermolysis bullosa simplex (EBS) with PLEC mutations, rare associations with dystrophic epidermolysis bullosa (DEB) secondary to COL7A1 defects have been documented [3,6]. Whereas classic Carmi syndrome involves JEB with cleavage through the lamina lucida and carries a poor prognosis, DEB results from type VII collagen (COL7A1) and anchoring fibril defects beneath the lamina densa [3,6,8,9]. Self-improving DEB, originally delineated as transient bullous dermolysis of the newborn, represents a distinct benign variant of DEB wherein severe neonatal skin fragility resolves spontaneously within early infancy without severe dystrophic scarring [8,10,11]. As clinical features of inherited bullous disorders can overlap in the neonatal period, definitively distinguishing between JEB, DEB, and self-improving variants generally requires a skin biopsy with immunofluorescence mapping, transmission electron microscopy, and molecular genetic

analysis [8-11].

The historical association of EB and CPA with rapid fatal outcomes had led clinicians to adopt guarded surgical interventions [1,5]. However, prompt operative correction and modern neonatology protocols have yielded improved survival [2,4]. We present a case of a neonate with type 1 CPA and intact skin at birth who developed self-limiting cutaneous bullae after successful pyloroplasty, with a self-improving course, complete resolution of blistering, and thriving development at long-term follow-up, a clinical trajectory presumptively consistent with self-improving DEB.

Case Presentation

A five-day-old male neonate presented with persistent non-bilious vomiting, feeding intolerance, reduced urine output, and fever. The patient was born at 40 weeks of gestation via lower-segment cesarean section, indicated for breech presentation, to a 24-year-old mother in a nonconsanguineous marriage. There was no family history of epidermolysis bullosa, congenital gastrointestinal malformations, or other blistering disorders. Antenatal ultrasonography at 21 weeks identified a mildly enlarged fetal stomach bubble, whereas third-trimester scanning at 37 weeks revealed polyhydramnios and asymmetric intrauterine growth restriction. The birth weight was 2,500 g, and resuscitation was not required at delivery. The baby gradually developed refusal of oral feeding along with non-bilious vomiting.

On admission at day five of life, the neonate was lethargic and moderately dehydrated with a sunken anterior fontanelle and dry oral mucosa. The abdominal examination revealed upper epigastric distension with visible gastric peristalsis and no palpable pyloric mass. There were no skin or mucosal lesions (oral, conjunctival, or perianal) at birth or upon admission. The plain abdominal radiography demonstrated a single dilated gastric air bubble with total absence of distal bowel gas (Figure *1*).

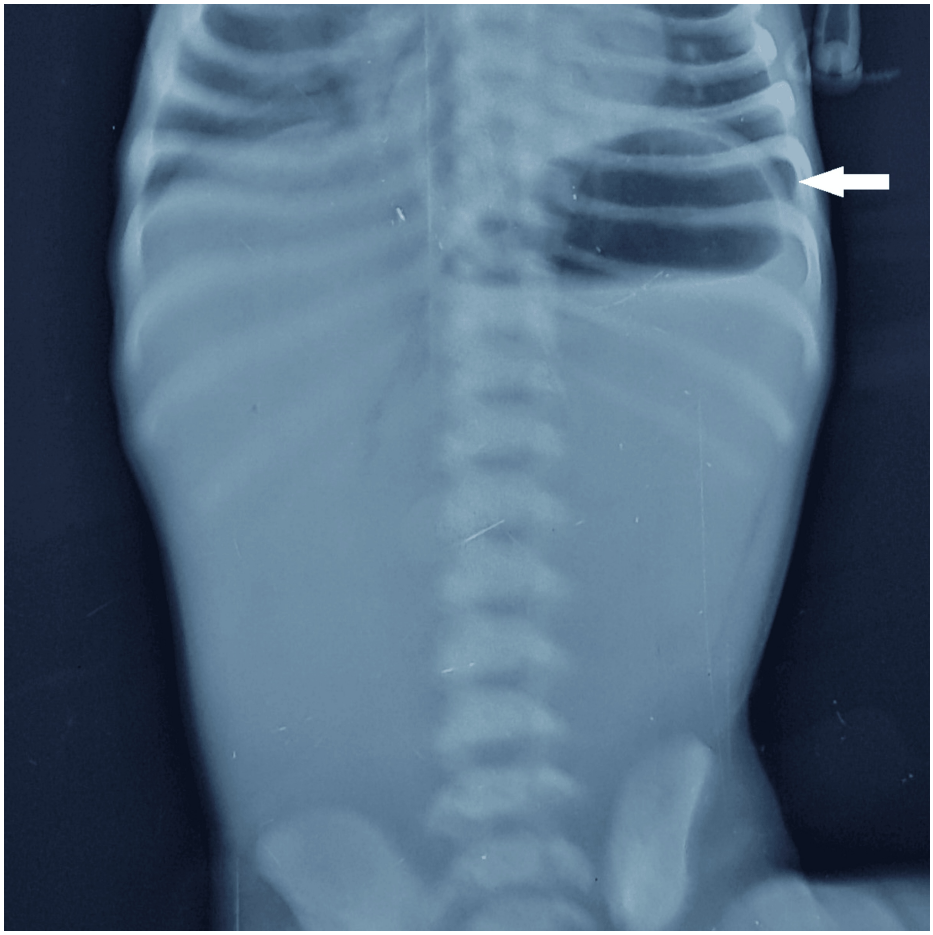


FIGURE 1: Plain abdominal radiograph obtained on day five of life showing a characteristic single dilated gastric air bubble (arrow) with complete absence of distal bowel gas, diagnostic of congenital pyloric atresia.

The abdominal ultrasound confirmed a massively distended stomach and an occlusive intraluminal pyloric mucosal diaphragm in the pyloric canal without duodenal communication or pyloric muscular hypertrophy, diagnostic of complete congenital gastric outlet obstruction and excluding infantile hypertrophic pyloric stenosis (IHPS). The echocardiography revealed normal cardiac anatomy with a 1-mm patent foramen ovale. The initial laboratory evaluation on admission demonstrated hemoconcentration and prerenal azotemia secondary to extracellular fluid depletion, without the classic hypochloremic metabolic alkalosis typically seen in IHPS (Table *1*).

Laboratory parameter	Patient value	Reference range, neonate	Unit of measurement
Hemoglobin	17.6	18.0-21.0	g/dL
Hematocrit	53.2	56.0-67.0	%
Urea	108	13-45	mg/dL
Creatinine	2.13	0.5-1.5	mg/dL
Lactate	3.3	0.5-2.0	mmol/L
Total bilirubin	15.20	4.0-8.0	mg/dL
Direct bilirubin	0.64	0.1-0.4	mg/dL
C-reactive protein	0.71	0.02-0.30	mg/dL

TABLE 1: Preoperative laboratory investigations.

Subsequent laboratory parameters were confirmed to be within established normal reference ranges for a five-day-old term neonate. Following stabilization and fluid rehydration, an exploratory laparotomy was performed on day seven of life via a right upper transverse incision. Direct surgical inspection corroborated the preoperative radiological findings, confirming a massively dilated stomach, thinned-out duodenum, a normal outer pyloric muscular layer without hypertrophy, and a thick intraluminal mucosal diaphragm completely occluding the pyloric canal, consistent with type 1 CPA. A longitudinal incision was made across the pylorus, the thick occlusive pyloric mucosal diaphragm was excised, distal bowel patency was verified with normal saline flushing, and a transverse two-layer Heineke-Mikulicz pyloroplasty was constructed. On the eighth postoperative day (day 15 of life), multiple spontaneous, tense, flaccid, fluid-filled bullae and superficial erosions appeared over friction-prone acral sites, specifically involving the left elbow, forearm, hand, and right foot (Figures *2*, *3*).



FIGURE 2: Multiple fluid-filled bullae over the left forearm and hand (arrows), occurring across friction-prone sites independent of intravenous cannulation, were noticed on the eighth postoperative day (day 15 of life).



FIGURE 3: An intact, fluid-filled bulla (star) and an adjacent superficial skin erosion (arrow) are seen over the right leg on the eighth postoperative day (day 15 of life).

No oral, conjunctival, or perianal mucosal blistering occurred during the acute episode. Intravenous fluid or medication extravasation injury was carefully excluded, as the bullae arose spontaneously across multiple non-cannulated acral sites. The diagnosis of epidermolysis bullosa was established on clinical grounds in consultation with the dermatology team, and microbiological testing of the blister fluid was therefore omitted as infectious etiologies were clinically excluded. Management focused on non-adhesive silicone dressings, topical mupirocin, and atraumatic nursing care, resulting in lesion resolution over the subsequent two weeks without scarring or milia. A transient, self-improving DEB phenotype was suspected based on the early onset of skin fragility and blistering that regressed within two weeks, the exclusion of extravasation injury, and the known association between epidermolysis bullosa and pyloric atresia, albeit representing a distinct subtype from this neonate. Although skin biopsy for immunofluorescence mapping and genetic multigene panel testing were recommended to confirm the diagnosis, both investigations were declined by the parents due to financial constraints, leaving the specific EB subtype as a presumptive clinical diagnosis. Low-volume oral feeding was initiated on postoperative day eight and gradually advanced until full enteral nutrition was established. There were no postoperative surgical complications. The neonate progressed well clinically and was subsequently discharged with a weight of 2,800 g.

At the one-month postoperative evaluation, the surgical scar was well-healed, the patient's weight had reached 3,200 g, and there was no recurrence of skin lesions. Follow-up at two years of age demonstrated normal overall growth and development, with residual postinflammatory hypopigmented patches over previously affected areas (Figure *4*). There were no active or new lesions, milia, or dystrophic scarring, and the nails were not dystrophic. The child had intermittent episodes of ocular pruritus during this period, which was evaluated by ophthalmology and attributed to conjunctivitis.



FIGURE 4: Residual postinflammatory hypopigmented skin patches over the left upper extremity at sites of previous blisters (arrows), with complete healing and absence of milia or dystrophic scarring observed at the two-year follow-up.

Discussion

The syndromic association of CPA with JEB, termed Carmi syndrome, represents an extraordinarily rare pathology encountered by pediatric surgeons [2,4]. While both CPA and EB most commonly occur as standalone or isolated conditions, clinicians must remain vigilant for their syndromic coexistence. The educational novelty of the present case lies in the initial presentation of pyloric obstruction with completely intact skin, masquerading as isolated CPA, and the subsequent postoperative emergence of a self-improving blistering phenotype that completely regressed [10].

From a diagnostic standpoint, CPA must be differentiated from IHPS, the most common surgical cause of non-bilious emesis in infancy [12]. Classically, IHPS presents later, at the second to 12th weeks of age, with progressive gastric outlet obstruction caused by hypertrophy of the pyloric musculature, frequently accompanied by hypochloremic, hypokalemic metabolic alkalosis [12]. In contrast, our patient exhibited hallmark features of CPA, namely an antenatal polyhydramnios detected at 37 weeks, symptom onset within the first five days of life, a classic single-bubble radiograph with complete absence of distal bowel gas, and sonographic demonstration of an occlusive intraluminal mucosal diaphragm without pyloric muscle thickening [1,2,12]. Metabolically, because neonates with CPA present within the first days of life, they typically manifest acute dehydration, hemoconcentration, and prerenal azotemia rather than the chronic metabolic alkalosis of IHPS, and surgical exploration on day seven definitively confirmed an occlusive intraluminal mucosal diaphragm rather than muscular hypertrophy. Similarly, when extremity bullae appeared on the eighth postoperative day (day 15 of life), iatrogenic intravenous fluid or medication extravasation was carefully ruled out. Although the antecubital fossa is a common venipuncture site, the spontaneous appearance of bullae across multiple non-cannulated, friction-prone acral surfaces confirmed mechanical skin fragility characteristic of EB. Moreover, there was no induration, necrosis, ulceration, or oozing as is usually seen in extravasation injuries [10,13].

In a 10-year systematic review of 49 patients with EB with pyloric atresia predominantly involving JEB

cohorts, it was found that 74% of neonates were born preterm, and 79% weighed under 2,500 g. Among the 43 patients who underwent surgical intervention for PA, 24 died postoperatively (55.8% operative mortality), almost exclusively secondary to septic shock, wound breakdown, and multiorgan failure [6]. Similarly, in a multicenter retrospective series of 152 EB patients, all four neonates diagnosed with PA had JEB, underwent Heineke-Mikulicz pyloroplasty, and ultimately died between four and eight months of age due to unrelenting sepsis [7]. It has been similarly documented in two neonates with type 1 CPA associated with epidermolysis bullosa who underwent diaphragm excision and pyloroplasty but succumbed at three and four months of age to intractable gastrointestinal fluid losses and severe dehydration, highlighting the historically high mortality seen in these complex cases [5]. Other authors have observed an 87.5% mortality rate among eight CPA-EB neonates, underscoring that cutaneous and systemic sepsis remains the overwhelming driver of mortality in classical junctional epidermolysis bullosa with pyloric atresia (JEB-PA) cohorts [1,4].

Type 1 CPA (occlusive pyloric mucosal web or diaphragm) represents the most common variant, documented in 57-60% of cases [1,2,5]. Excision of the occluding mucosal diaphragm coupled with a Heineke-Mikulicz pyloroplasty is the operation of choice [1,4,5]. Intraoperative verification of distal intestinal patency by intraluminal catheterization or saline flushing is mandatory, as associated distal atresias occur in up to 25% of patients [1,4]. While gastrojejunostomy has been described, it carries a high risk of marginal ulceration and stomal stenosis in neonates and should be avoided [1,2]. In our patient, excision of the occluding pyloric mucosal diaphragm with pyloroplasty on day seven of life effectively re-established gastrointestinal continuity without complications [1,2,5].

The vast majority of epidermolysis bullosa with pyloric atresia (EB-PA) cases are categorized as JEB caused by ITGB4 or ITGA6 mutations, or epidermolysis bullosa simplex (EBS) caused by PLEC mutations [3,6]. Association of CPA with DEB is exceedingly rare, with only five prior ultrastructurally confirmed cases worldwide having been documented [3]. In the present case, blistering manifested on the eighth postoperative day (day 15 of life), predominantly involving friction-prone acral regions [3,10]. Instead of following the lethal downhill course typical of JEB-PA, the patient exhibited a distinctive benign clinical course consistent with self-improving DEB, historically described as transient bullous dermolysis of the newborn [8,10]. In ultrastructurally confirmed cases of this entity, blister formation occurs beneath the lamina densa due to transient collagenolysis and anchoring fibril disruption, accompanied by distinctive dilatation of the rough endoplasmic reticulum containing electron-dense stellate bodies within basal keratinocytes. It was observed that new blistering ceases spontaneously within the first few months of life, healing rapidly with postinflammatory hypopigmentation rather than dystrophic scarring or milia, a benign clinical course that mirrors our patient's two-year outcome [8].

This self-limiting disease pattern has been further corroborated in a study on four cases of bullous dermolysis of the newborn, which highlighted that cutaneous blister activity is characteristically restricted to early infancy, typically resolving between three and seven months of age completely. The study also noted that while immunofluorescence mapping classically reveals intraepidermal granular deposits of type VII collagen that subsequently normalize to a linear dermoepidermal junction pattern as lesions resolve, high diagnostic costs often preclude routine genetic sequencing [11]. In molecularly characterized cohorts, self-improving DEB is driven by COL7A1 glycine substitutions causing temporary intracytoplasmic retention of type VII collagen within basal keratinocytes; as epidermal maturation progresses, collagen processing normalizes, leading to spontaneous clinical resolution [10,11]. A key limitation in our patient is that we could not perform skin biopsy for immunofluorescence mapping, electron microscopy, or molecular genetic testing due to financial constraints [8,9,11]. While spontaneous cessation of blistering and absence of severe dystrophic scarring at the two-year follow-up confirm a self-improving EB clinical course, they cannot definitively prove the molecular subtype in the absence of immunofluorescence mapping or molecular genetic testing; thus, the diagnosis of self-improving DEB in our patient remains presumptive [8-11]. Concordantly, our patient demonstrated complete cessation of blistering with age, leaving only residual hypopigmented patches [8,10,11].

Conclusions

The coexistence of congenital pyloric atresia and epidermolysis bullosa represents a complex neonatal emergency that has historically carried high mortality, particularly in classical JEB-PA cohorts. While the favorable outcome of our single patient with a self-limiting blistering course cannot be generalized to severe JEB-PA populations, the documentation of a self-improving blistering phenotype underscores that the association of CPA and EB encompasses a heterogeneous clinical spectrum, and surgical repair with rigorous skin protection should be pursued in stable neonates. Pyloric atresia has been most commonly observed with JEB, with associated high mortality, whereas DEB has been reported with pyloric atresia in around five cases. Self-improving DEB is a rare subtype of DEB previously known as transient bullous dermolysis of the newborn, characterized by early-onset skin fragility and blistering that usually regresses within the first few months to years of life. Although definitive subtyping as self-improving DEB could not be confirmed via immunofluorescence mapping or molecular genetic testing due to financial constraints, longitudinal follow-up over two years confirmed a benign, self-improving phenotype with complete cessation of blistering. This case represents a rare clinical presentation of type 1 CPA associated with a self-improving EB phenotype presumptively consistent with self-improving DEB.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

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Acquisition, analysis, or interpretation of data: Lalit Parida, Aditi Parida, Jagdish Prasad Sahoo, Asha MN

Drafting of the manuscript: Lalit Parida, Aditi Parida, Jagdish Prasad Sahoo, Asha MN

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Supervision: Lalit Parida, Aditi Parida, Jagdish Prasad Sahoo, Asha MN

Disclosures

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