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Case Report of a Rare Anomaly: Congenital Pyloric Atresia Associated with Bart Syndrome

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INTRODUCTION

Pyloric atresia (PA) is an extremely rare congenital anomaly (with an incidence of 1 in 100,000 live births).
It carries a high mortality rate when associated with epidermolysis bullosa (EB), a genodermatoses characterized by skin fragility and trauma-induced blister formation. Bart's syndrome is an autosomal dominant disease characterized by EB, aplasia cutis (AC), and nail abnormalities. To the best of our knowledge, this is the first case of Bart's syndrome associated with PA reported in Malaysia.

CASE REPORT

We present a case of a term newborn male, prenatal scans showed polyhydramnios and a dilated stomach bubble. Postnatally, baby had abdominal distension with copious non bilious aspirates. Plain radiograph identified a 'single bubble' with absence of gas distally, while bedside ultrasound ruled out intestinal malrotation. Correlation between PA and EB (junctional type) was made when trivial trauma caused extensive skin erosions and eruption of blisters, diagnosis confirmed with skin biopsy.

Intraoperatively, a PA type 2 repair was done with a grastroduodenostomy and transanastomotic tube insertion. Special care was taken to prevent further trauma to the skin during and after surgery. Postoperative, patient had achieved full tube feeding. Oral feeding was not commenced in view of worsening oral ulcers. The skin lesions were meticulously managed by our paediatric dermatologist. Patient however had bouts of gram positive cocci sepsis and eventually succumbed to death at three months of age.

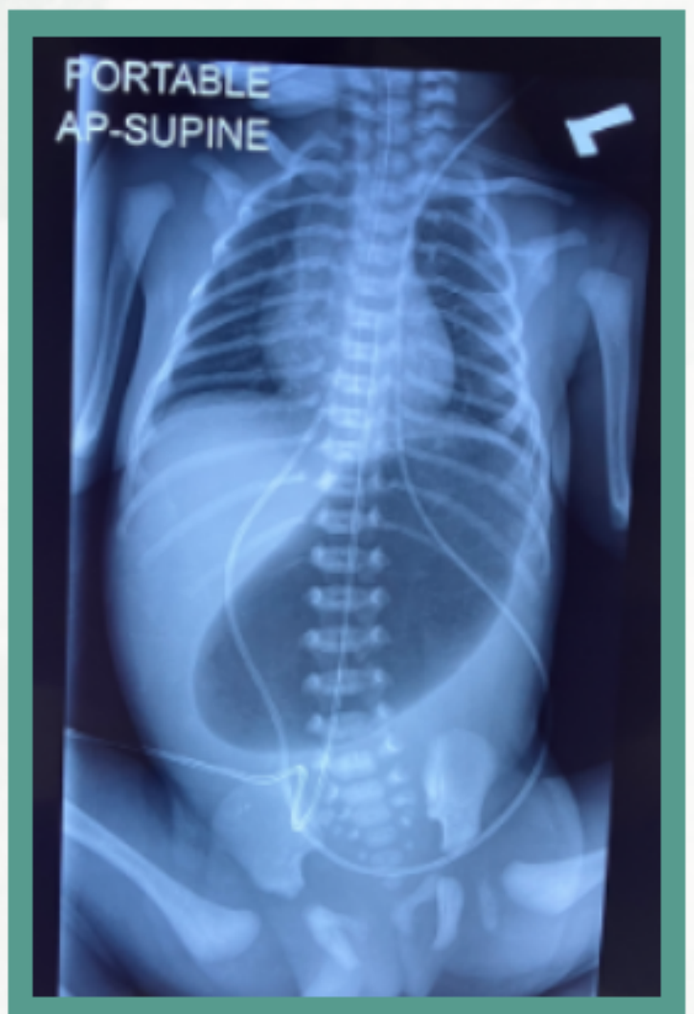

Figure 1. Plain radiograph demonstrating a hugely dilated stomach ('single bubble sign') with absence of distal gas.

DISCUSSION

Surgical correction in isolated PA confers a good prognosis. However mortality rate is high when diagnosis of EB is also present. [1,2] Immature neonate immune system combined with frequent and extensive loss of protective skin barrier predispose patient with EB to risk of infection and electrolyte imbalances from fluid loss.[3]

Special care as to not induce trauma to the skin had been taken during preparation and transfer for surgery. Bipolar diathermy was preferably used when possible to avoid friction from placement of diathermy pads. A central venous catheter was placed for parenteral nutrition and to reduce repeated skin pricking for intravenous access. Feeding was established via orogastric tube as the patient had developed intraoral ulcers that had made it difficult to start oral feeds. Wound care proved to be most challenging, with use of non adhesive pad and two-sided wound contact layer dressings to promote wound healing. Despite the exhausted efforts in wound management, patient remained susceptible to wound infection and severe septicemia that led to mortality.

Figure 2.
Blister formation (ruptured) on the right forehead (A) and right thigh (D).
Lack of skin on the left thumb (B) and bilateral elbow joints (C).

CONCLUSION

PA with EB is an extremely rare and fatal disease. Unfamiliarity combined with high mortality rate requires coordinated care by multidisciplinary team of paediatric surgeon, neonatologist, paediatric dermatologist, paediatric anaesthesiologist, and trained neonatal intensive care unit (NICU) staff to ensure favorable outcome.

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