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# CONGENITAL PYLORIC ATRESIA AND EPIDERMOLYSIS BULLOSA

Pages with reference to book, From 254 To 256

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## INTRODUCTION

Congenital pyloric atresia with epidermolysis bullosa is recognized as a rare but distinct entity which is uniformly fatal<sup>1,2</sup>. Until 1990 only 20 cases have been reported in the English literature<sup>3,4</sup>. We present an additional case managed at the Aga Khan University Hospital to demonstrate the difficulties encountered in the management of such infants.

## CASE PRESENTATION

SBS, a 1700 gin male was the product of a full term pregnancy in a 29 year old healthy woman. The pregnancy was complicated by polyhydramnios but was otherwise uneventful. The parents were not blood relatives, but the paternal grand mother and maternal grand father were first cousins. Five siblings, all females, were normal and the family history was unremarkable. Soon after birth, the baby developed vomiting, respiratory distress and was transferred to the Aga Khan University Hospital. On admission the baby was moderately dehydrated and tachypneic. The abdomen was fiat and non-tender. Bullous lesions were noted on both upper lips. A radiograph of the abdomen revealed gas in the stomach in an otherwise gasless abdomen (Figure 1).

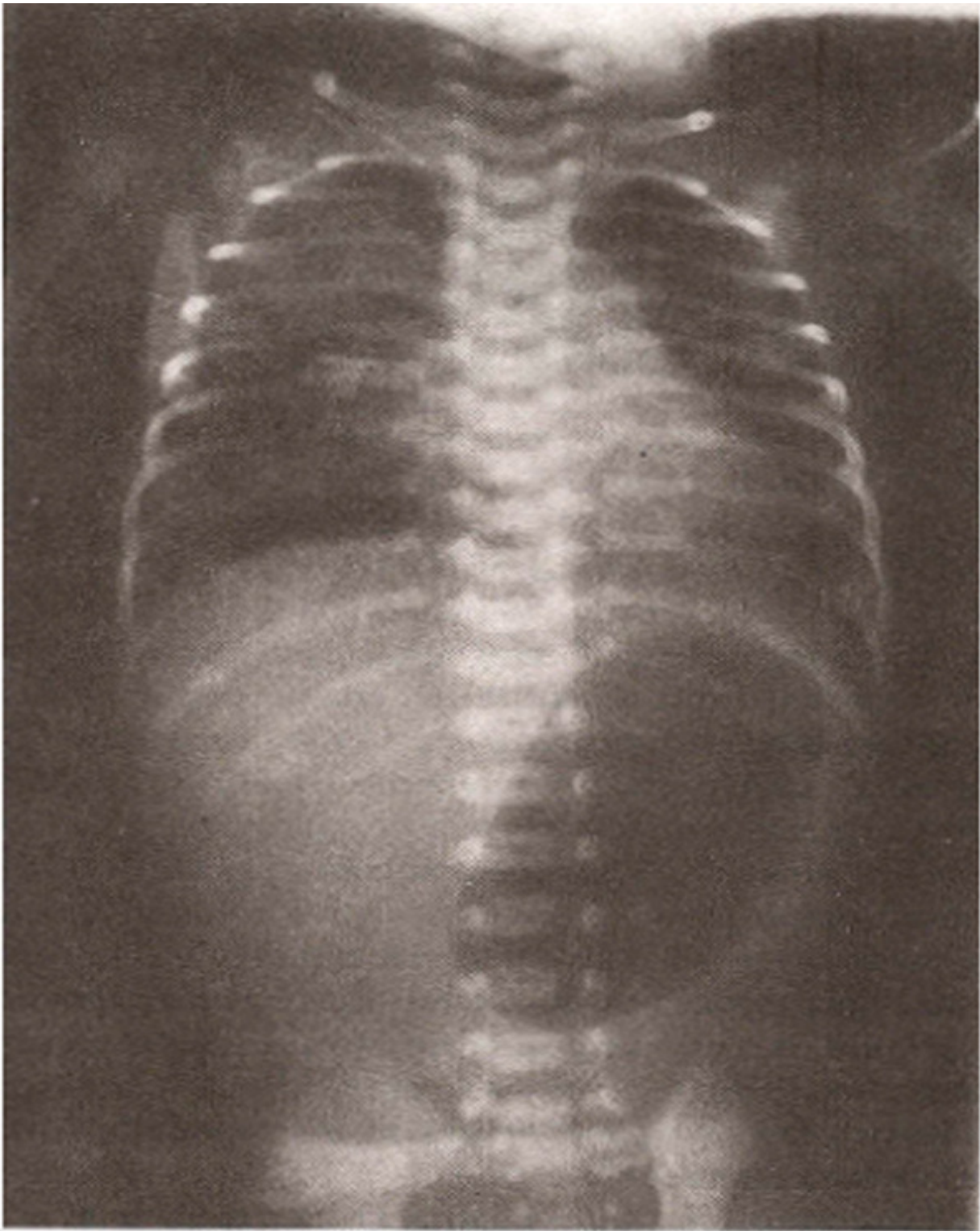


Figure 1. Appearance of abdominal roentgenogram in pyloric atresia. Note the large gastric air bubble without distal intestinal air.

A limited barium meal confirmed complete gastric outlet obstruction (Figure 2),

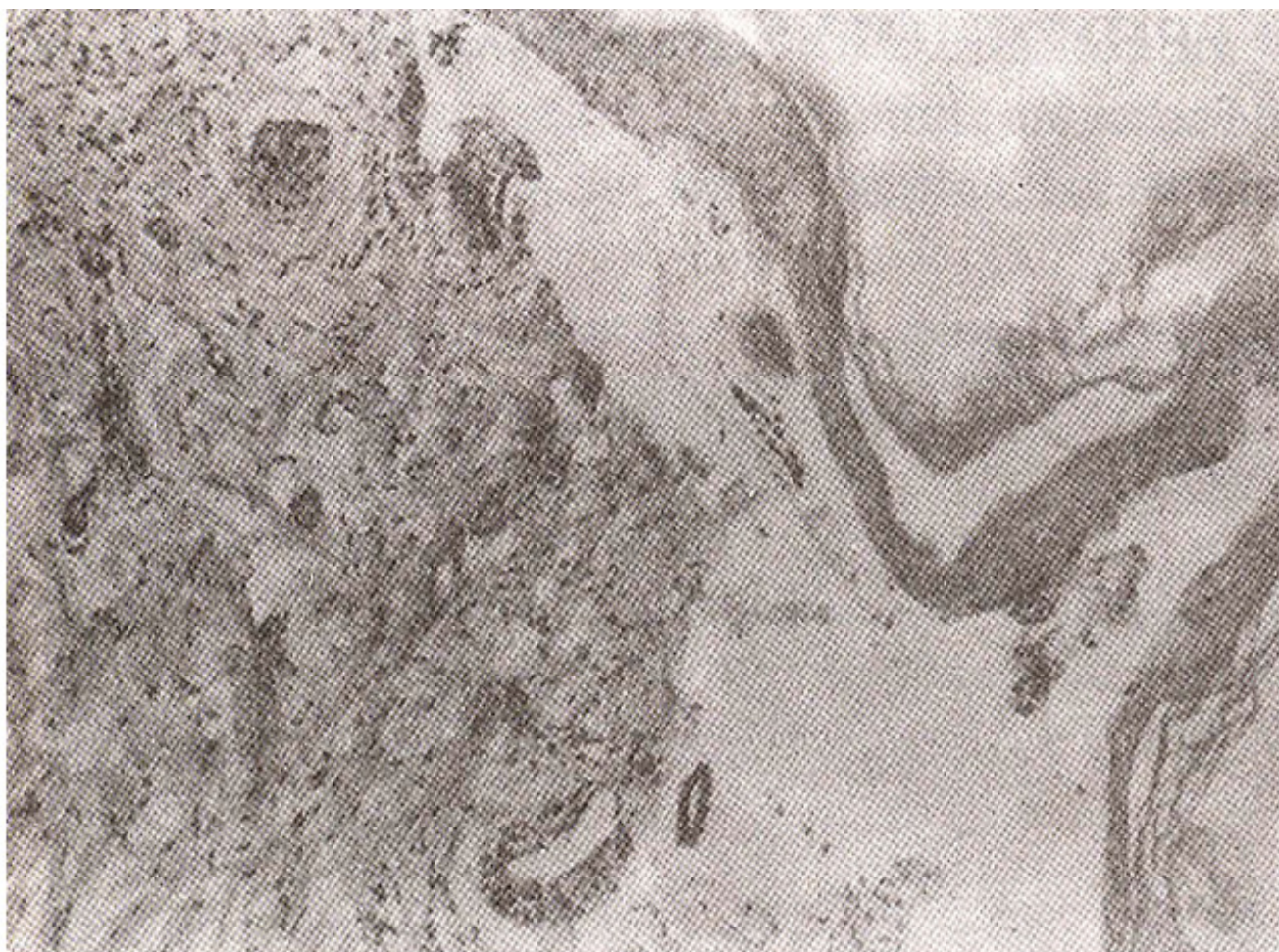




**Figure 2. Upper G.I. showing complete obstruction at the pylorus.**

an exploratory laparotomy confirmed pyloric atresia and a gastrojejunostomy and feeding jejunostomy were performed. Post-operatively, fresh crops of blisters continued to appear notably on the skin of the limbs, lumbosacral region and on areas where adhesive tape was applied. A skin biopsy sent for routine microscopy was consistent with the diagnosis of epidermolysis bullosa (Figure 3).





**Figure 3. Subepidermal bulla with focal vacuolization and degeneration of basal cells (x200).**

The infant continued to deteriorate with worsening of the blisters, and subsequent bacterial and fungal sepsis compounded by malnutrition due to: feeding difficulties and plasma losses from the blisters. He died of progressive sepsis and disseminated intravascular coagulation (DIC) at 2 months of age.

## **DISCUSSION**

The association of congenital pyloric atresia and epidermolysis bullosa although rare, occurs often enough to have been recognized as a "syndrome"<sup>1-4</sup>. Congenital gastrointestinal atresias occur in one in 10,000 births, with pyloric atresia constituting only 1% of all reported cases<sup>1,4,5</sup>. Pyloric atresia may consist of a mucosal diaphragm, a solid cord or a thin fibrous membrane and is thought to be the result of mechanical or vascular injury to the fetal intestine. A familial occurrence is reported and an autosomal recessive transmission has been suspected<sup>1,4,6,7</sup>. A gastroduodenostomy or a gastrojejunostomy is usually advocated for management<sup>6</sup>. Epidermolysis bullosa forms a heterogeneous group of inherited disorders, characterized by extreme fragility of the skin and bullae formation<sup>8</sup>. The prevalence of the recessive varieties has been cited to be 1:45,000<sup>9</sup>. The condition is classified into three major groups, simplex, junctional and dystrophic, based on the electron microscopic picture<sup>8</sup>. The junctional and dystrophic varieties are severe and almost always lethal. In addition to the cutaneous manifestations, there can be involvement of the oral and oesophageal mucosa and the genitourinary and respiratory tracts. The bullae heal with or without scarring. Severe growth retardation secondary to

malnutrition and infection usually cause death in infancy. At present there is no successful treatment for epidermolysis bullosa, although Phenytoin is reported to be useful<sup>10</sup>. The simultaneous occurrence of the two conditions is interesting. Although some have speculated that the pyloric atresia may be secondary to the mucosal lesions of epidermolysis bullosa, most believe that the two entities are associated in an autosomal recessive syndrome, caused by the homogenous state of a single mutant pleiotropic gene<sup>3,5</sup>. The consanguinity of the grandparents of our patient is consistent with an autosomal recessive inheritance. The association of junctional and dystrophic variants of epidermolysis bullosa and congenital pyloric atresia is lethal, with only one child reported to have survived until 11 months of age<sup>5</sup>. As demonstrated in our case and also reported in English literature, the surgical correction of the associated pyloric atresia does not appear to influence the overall survival. We believe that the treatment of this disorder should be symptomatic and supportive only. Nutritional support using total parenteral nutrition has been advised but in countries like Pakistan such a facility is expensive and available to only a small percentage of the population<sup>5,11</sup>. A feeding jejunostomy may provide an alternative route for nutrition and allow early discharge of the patient. Although a prenatal diagnosis is possible by electron microscopic examination of a fetal skin biopsy taken as early as 18 weeks of gestation,<sup>12,13</sup> but this procedure is not available in Pakistan. Prevention therefore must be sought by means of genetic counselling as the parents of a child with pyloric atresia epidermolysis bullosa syndrome have a 1:4 risk of recurrence in subsequent off-springs<sup>4,5,7</sup>. The presence of epidermolysis bullosa in new born infant should alert the pediatrician to the possibility of a co-existing pyloric atresia.

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