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EPIDERMOLYSIS BULLOSA ASSOCIATED WITH PYLORIC OBSTRUCTION*

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Several recent reports have called attention to the association of epidermolysis bullosa and pyloric obstruction[1]. We present two newborns with this combination and additional abnormalities.

Case Reports

Case 1

A two-hour-old male infant was admitted to our hospital because of skin lesions and dysmorphic features. He was born to a 26-year-old gravida 7, para 2 mother following a premature spontaneous vaginal delivery. Polyhydramnios was noted. The mother's first two pregnancies were uneventful, the following three ended in spontaneous abortion in the first trimester, and the product of the sixth pregnancy was a full-term stillborn.

The parents were first cousins. The infant appeared premature with an AGA of 34-36 weeks. He weighed 1280 g and was markedly growth retarded. Superficial skin erosions and blisters were noted all over his body but deep, symmetrical, sharply demarcated skin defects were confined to the scalp, face and neck. He had a prominent forehead and nasal bridge, flat vertex and receding chin. Both auricules were malformed and fixed to the scalp. Other abnormal findings included a high-arched palate, obstructed nostrils, single umbilical artery, lumbosacral meningocele, third degree hypospadias, malformed feet with prominent calcaneus and a missing right small toe. Radiologic findings revealed a deformed and lacunar skull, spina bifida at L 5, and a single huge gas bubble on the

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upper abdomen. A barium swallow disclosed a filling defect at the antral area and no passage of barium beyond the stomach (Fig. 1). He was given supportive care but died of aspiration at two days of age.

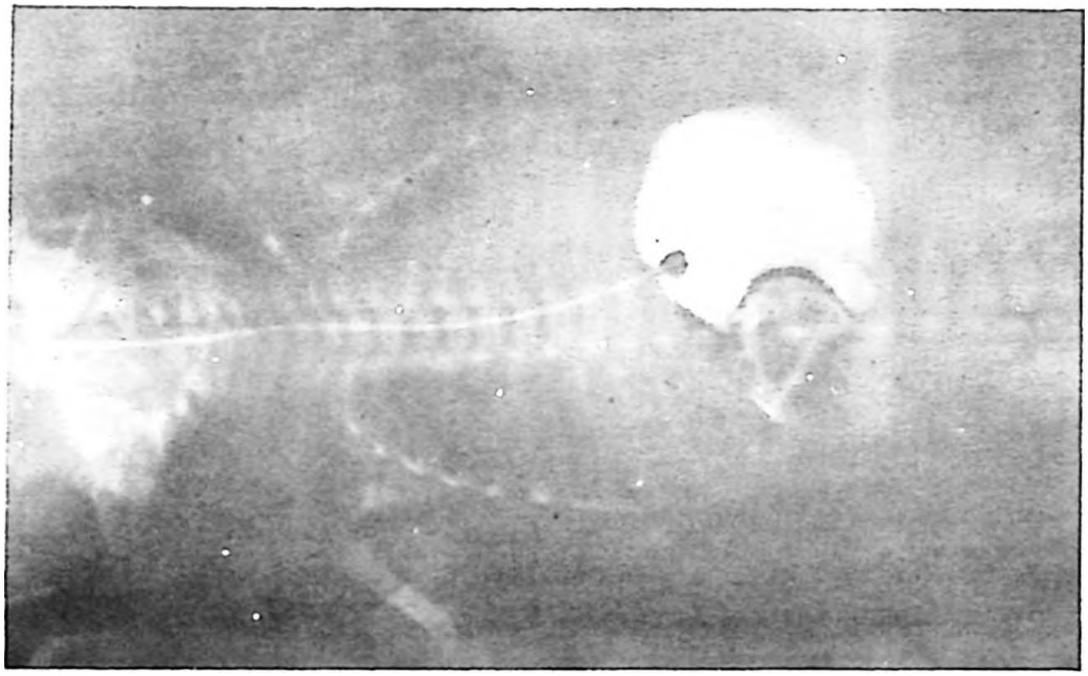

Fig. 1: Radiologic appearance of antral filling defect and gastric outlet obstruction in Case No. 1.

At postmortem examination pyloric lumen was present but the pylorus and proximal part of the duodenum had prolapsed into the stomach. No histologic abnormalities were noted on light microscopy excluding congested serosal vessels. Light microscopic examination of the skin sections from relatively normal appearing areas disclosed normal dermal adnexal structures and dermal epidermal separation above the dermis, suggesting junctional epidermolysis bullosa. Sections from the defective areas resembled histologic findings of aplasia cutis with the absence of dermal adnexal structures and a thin adherent overlying epidermis. The findings in the brain, liver and kidneys were compatible with prematurity and were otherwise unremarkable.

Case 2

A five-hour-old female infant was born to a 21-year-old mother at 36 weeks of gestation complicated with polyhydramnios and premature rupture of membranes. The parents were first cousins. Physical examination revealed a borderline premature infant with marked growth retardation and respiratory distress. Extensive skin defects were observed on the face, neck, upper and lower

extremities and over the periumbilical area (Fig. 2). Excessive oropharyngeal secretions, bilateral corneal opacities and an epigastric smooth intraabdominal mass were other unusual findings.

Fig. 2: Skin defects on lower extremities and at periumbilical area in Case No. 2.

Laboratory findings: hemoglobin 9.7 g/dl, WBC 7000/mm^3 (with 40% atypical mononuclear cells and 35% nucleated red blood cells). There was no blood group incompatibility. Coombs test, and serologic tests for toxoplasma and syphilis were negative. Urinalysis, CSF, and the blood chemistry were within normal limits. X-rays showed an airless abdomen and a faint linear calcification at the left upper abdomen. Attempts to insert a nasogastric tube were unsuccesful due to resistance in the lower pharynx. The infant was suspected of having esophageal atresia with epidermolysis bullosa and died after a prolonged apneic episode three hours after admission.

Postmortem examination revealed a patent esophagus but the pylorus was atretic and the mass palpated was found to be a fluid filled stomach. Microscopically no lumen or mucosal structures were observed but marked disarrangement of smooth muscle fibers was present in the pyloric area. The colon was underdeveloped and increased extramedullary hematopoiesis was evident in the enlarged liver and spleen. Histologic findings of the skin were entirely similar to the findings in the first case.

Discussion

Epidermolysis bullosa with pyloric atresia is an association which had previously been described. Fourteen cases have been reported since 1979. Skin involvement in these infants appears to be either superficial blisters with random distribution[1-3] or sharply demarcated symmetrical skin defects[4,5]. Some of the cases, including ours, had both. Pyloric atresia has been noted shortly after birth in all cases but one[6], who developed severe postnatal stenosis. Our patients had some unreported findings such as gastric outlet obstruction, cranio-facial malformations, meningocele, a single umbilical artery, corneal clouding and Coombs negative hemolytic anemia. Prolapse of the pylorus into the gastric lumen in our first case was puzzling and suggests a pyloric functional disturbance as well.

In previous reports, authors have discussed the possibility of inheriting these two genetic conditions simultaneously and investigated the type of epidermolysis bullosa in depth. Both scarring and nonscarring forms have been reported.

Skin defects in these infants were thought to be scars secondary to intrauterine trauma. The same mechanism was suggested for pyloric atresia by some authors while others proposed inheritance of both conditions simultaneously.

Although epidermolysis bullosa is a constant finding, the type of this disease does not seem to explain the presence of skin defects and other abnormalities. Furthermore defective skin areas probably do not represent true scars. With lacking connective tissue, these lesions histologically resemble more the findings in aplasia cutis congenita.

Additional developmental anomalies in our cases widen the spectrum of this anomaly complex. The presentation of these two infants from the same area who were born four months apart to mothers unrelated was quite interesting and led us to consider the possibility of a similar superimposed adverse intrauterine effect in these epidermolytic fetuses. Another possibility is the inheritance of a wider range of anomalies with defective close gene locations as previously suggested[2].

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