Epidermolysis Bullosa – Pyloric Atresia Syndrome

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Epidermolysis bullosa (EB) is an uncommon congenital blistering disorder of the skin. Atresia of the pyloric end of stomach is also rare, constituting only 1% of all atresias of the gastro-intestinal tract. The association of pyloric atresia and epidermolysis bullosa is so rare that only twelve cases have been previously reported. Here we report a preterm boy with epidermolysis bullosa and associated pyloric atresia.

CASE REPORT

A boy weighing 1900 g was born to a 32 year old fourth gravida woman at 32 weeks of gestation. There was no associated polyhydramnios. At birth, baby had partial absence of skin in the upper and lower limbs with large blisters. The skin peeled off on gentle pressure. Peeling off skin was prominent on the nose, cheeks; both ears, thighs and the gluteal region. Hair and nails were normal. Blood culture was sterile. VDRL of both parents and baby were non-reactive. Baby died on the second postnatal day inspite of active management.

Her first baby was also a preterm male infant with similar clinical features but died at 12 hours of age. The second and third siblings were normal. Father was 36 years old and there was no history of consanguinity in the family.

Autopsy Findings. Extensive peeling of skin was seen on both the upper and lower limbs exposing the underlying vessels and muscles. Internal examination showed early evidence of bronchopneumonia in the lungs. The bronchi and bronchioles showed separation of the lining epithelium in areas. The heart and great vessels were anatomically normal. The stomach appeared distended. The whole of pyloric area was atretic, (Fig. 1) cord like and could not be opened up. The oesophagus showed separation of the lining epithelium. The kidneys showed separation of the transitional epithelium of the pelvis.

Histopathology of skin showed hyperkeratosis with focal keratotic plugging. Sections from the junction of the bullous lesion with the rest of the skin showed subepidermal/junctional bullae (Fig 2). The epidermis of the bulla showed thinning with moderate loss of prickle cell layer. The dermis and deeper adipose tissues were normal. The dermal appendages showed partial separation from the basement membrane in areas.

DISCUSSION

Epidermolysis bullosa is a heterogeneous group of congenital hereditary blistering disorders, characterised by vesiculo-bullous lesions, appearing either in utero or during
Fig. 1: Pylorus atrertic and cord like (indicated by arrows).

Fig. 2: Skin histopathology showing junctional bullae (arrow).