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Antenatal Diagnosis of Epidermolysis Bullosa – Pyloric Atresia - A Rare Case Report with A New Radiological Sign

Dr. Ramya Konareddy[1], Dr. Pramod Setty J[2], Dr. Jeevika MU[3]
[1](Post Graduate Resident, Department Of Radio-Diagnosis ,JJM Medical College , Davangere , India)
[2](Professor , Department Of Radio-Diagnosis ,JJM Medical College , Davangere , India)
[3](Professor , Department Of Radio-Diagnosis ,JJM Medical College , Davangere , India)

Abstract : *Association of congenital pyloric atresia with epidermolysis bullosa (EB-PA) is a rare autosomal recessive disorder. We report a case of EBPA which was suspected on antenatal ultrasound examination and confirmed later when baby delivered.*
Keywords: *epidermolysis bullosa, congenital pyloric atresia, hydronephrosis*

I. Introduction

Pyloric atresia (PA) is a rare malformation which is estimated to be responsible for less than 1% of gastrointestinal atresia with an incidence of 1 in 100,000 live births[1]. Epidermolysis bullosa (EB) represents a heterogeneous group of rare inherited disorders characterised by fragility of the skin and mucous membrane, which manifest by spontaneous or post-traumatic blister formation and skin peeling. The incidence of EB is 1 case/50 000–500 000 live-births[2]. Evidence suggests that EB-PA association is a distinct entity, referred to as the EB –PA syndrome[3]. Swinbure and Kohler first described the association of PA and EB in 1968[4].

II. Case History And Imaging Findings

G3P2L2 female presented with 8 months of amenorrhea and pain abdomen. Patient was referred for ultrasonography for fetal well being. No previous scans were done. History of second degree consanguinity was present. Previous two pregnancies were uneventful and babies were hail and healthy. Antenatal ultrasonography showed fetus corresponding to 30 weeks gestation with single dilated stomach bubble, with nonvisualisation of distal bowel loops, bilateral hydronephrosis and a thin bullous lesion on the chest wall and echogenic amniotic fluid (snow flake sign). Patient progressed in labour and delivered a preterm baby with marked epidermolysis bullosa lesions and areas of aplasia cutis and postnatal usg showed pyloric atresia features and bilateral hydroureteronephrosis. Post natal erect radiograph showed single bubble sign. Baby died 1 day after delivery. Autopsy was not done as patient attenders were not ready to give consent.

III. Figures

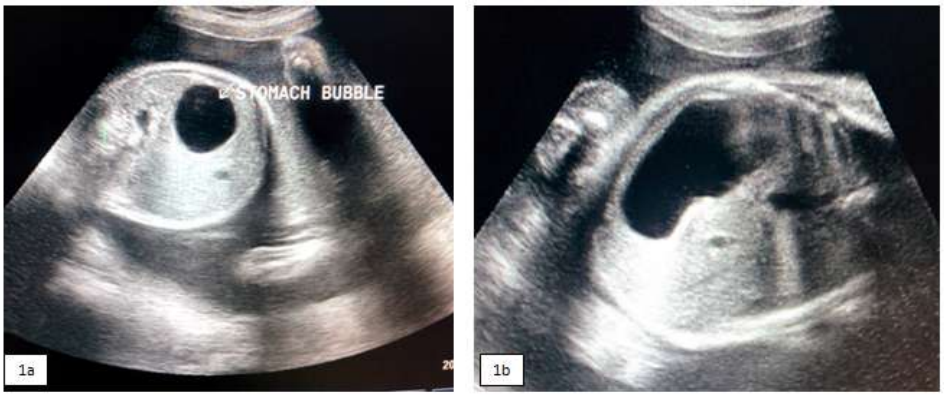

Fig1a and 1b : Antenatal ultrasonography images showing Dilated stomach bubble with paucity of bowel loops in the fetus – Pyloric Atresia

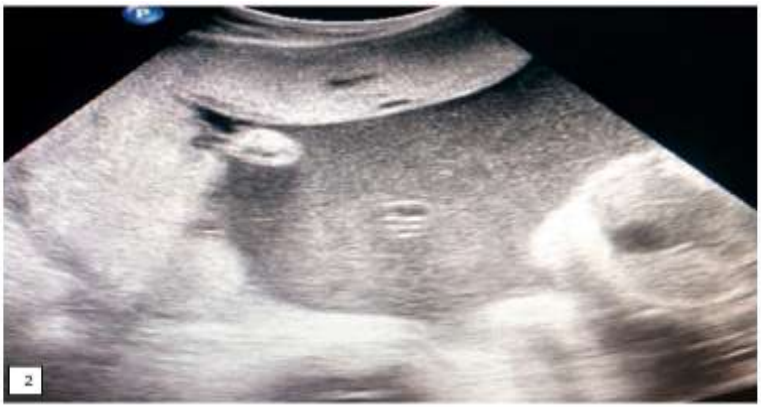

Fig2: Antenatal ultrasonography images showing thick echogenic amniotic fluid – SNOW FLAKE SIGN

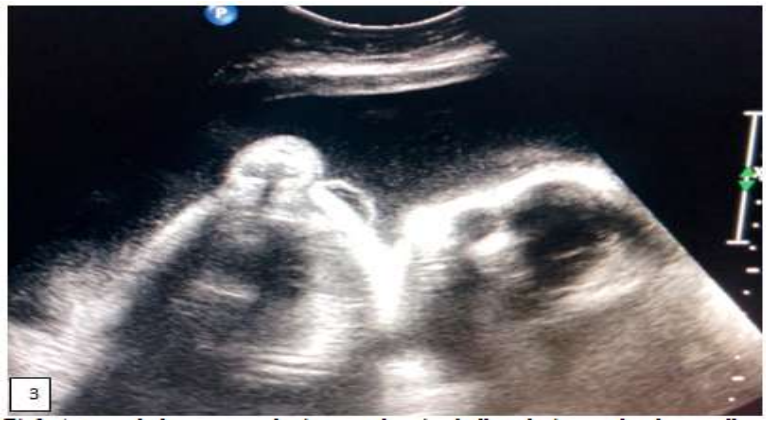

Fig3: Antenatal ultrasonography images showing bullous lesion on the chest wall

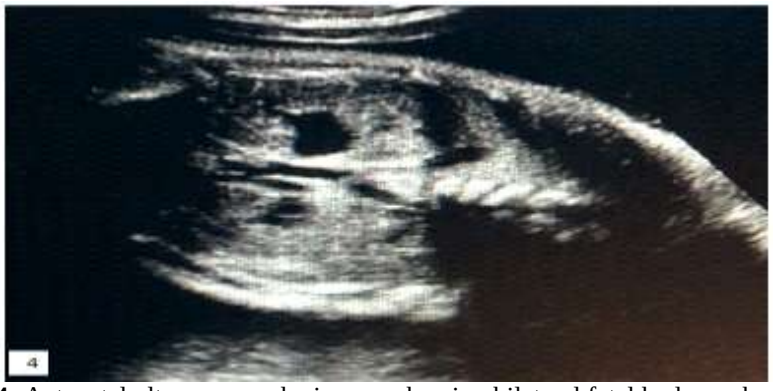

Fig4: Antenatal ultrasonography images showing bilateral fetal hydronephrosis

Fig 5a and 5b: Delivered preterm baby showing features of epidermolysis bullosa , aplasia cutis and nail changes

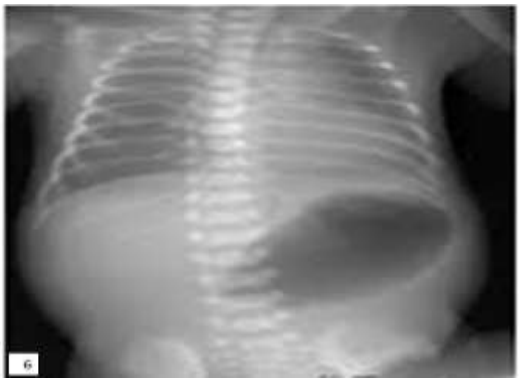

Fig6: Post delivery upright radiograph showing single bubble sign – pyloric atresia

IV. Discussion

Epidermolysis bullosa-pyloric atresia (EB-PA) represents a syndromic association of skin fragility and congenital gastrointestinal atresia, most frequently pyloric, although duodenal atresia with skinfragility has also been reported[5]. The course of EB-PA is usually severe and often lethal in the neonatal period. Most affected children succumb as neonates; those who survive may have severe blistering with formation of granulation tissue on the skin around the mouth, nose, fingers, and toes, and internally around the trachea.

Numerous subtypes of EB are described and divided into three major groups: EB simplex(EBS), dystrophic EB(DEB) and junctional EB (JEB). Association of JEB with pyloric atresia is termed as Carmi syndrome[6]. Patients with Carmi syndrome can also present with erosions and subepithelial cleavage in the respiratory, gastrointestinal and urinary tracts [7]. Other features include congenital localized absence of skin (aplasia cutis congenita) affecting the extremities and/or head, milia, nail dystrophy, scarring alopecia, hypotrichosis, contractures, and dilated cardiomyopathy[8]. Dang et al., in their recent study published in 2008, reported three cases and reviewed 46 cases of JEB-PA published in the literature. In addition to PA (n=49), other commonly reported complications in these patients included nail dystrophy (n=7), enamel hypoplasia (n=4), aplasia cutis congenital or congenital localized absence of skin (n=6), eye involvement (n=4), ear or nose hypoplasia or atrophy (n=4), urinary tract involvement (n=8), and respiratory involvement (n=5)[9].

The reported sonographic signs of EB include gastric dilatation associated with polyhydramnios as an expression of pyloric atresia, hydronephrosis and urethral stenosis, ear and nose deformities, and the snowflake sign, which may be a sign of in utero skin denudation syndromes[10].

In 1990, Meizner and Carmi described the Snow Flake sign, an echogenic appearance of the amniotic fluid during the second trimester of pregnancy associated with EB[11]. In all patients with snowflake amniotic fluid, Carmisyndrome should be suspected[6].

In literature, visualization of bullous lesion antenatally is not described to the extent of our knowledge and can be considered as a new antenatal sign.

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