Aplasia cutis congenita, group 5 without fetus papyraceus in two newborns

Sir,

Aplasia cutis congenita is an uncommon disorder characterized by the congenital absence of a portion of the skin, the most common presentation being a solitary lesion on the vertex of the scalp, just lateral to the midline (70%). Lesions are usually non-inflammatory, well-demarcated and have a variable appearance depending on the size (0.5–10 cm), extent, depth and degree of healing and scarring.[1,2]

The various causative pathophysiological mechanisms include genetic factors, intrauterine trauma, fetus papyraceus, intrauterine infections by varicella or herpes viruses, drugs including methimazole, carbimazole and misoprostol, feto-fetal transfusion, early rupture of amniotic membrane forming amniotic bands and abnormal elastic fiber biomechanical forces.[1,2] Truncal aplasia cutis congenita is usually associated with fetus papyraceus, classified as group 5. We report a rare presentation of aplasia cutis congenita, group 5 in two newborns without any history of twin gestation/fetus papyraceus.

Case 1

A 2-day-old healthy male child with a birth weight of 2.9 kg, delivered by full-term normal vaginal delivery to a multigravida mother, presented with congenital absence of skin on bilateral flanks. Ultrasonography done at 18 and 32 weeks antenatally had revealed a single live fetus with normal parameters and with no obvious congenital malformations. On examination, the lesions had a gelatinous appearance with a glistening red base and visible capillaries covered with a thin transparent membrane. They were 6.3 cm × 4.2 cm, well-defined and symmetrical, present on bilateral flanks joined with a linear streak [Figure 1a].

Case 2

A newborn healthy male child, weighing 2.8 kg. delivered by full-term normal vaginal delivery, presented with absence of the skin on the trunk, arms and legs. His mother had a history of typhoid fever at 6 weeks of gestation which was managed conservatively; the antenatal period was otherwise uneventful. Ultrasonography at 18 and 28 weeks antenatally revealed a single live fetus with healthy parameters and no twin gestation was noted. On examination, the lesions were well-demarcated, symmetrical, linear/elliptical and stellate. They were covered with a thin, gelatinous, shiny membrane and were present on both flanks, upper arms and thighs, distributed in a H-shaped pattern measuring 7.1 cm × 4.4 cm [Figure 2a].

There was no history of any intrauterine infection/trauma or exposure to any teratogenic drug in both cases. Scalp and nail examination, radiological examination and ultrasonography revealed no abnormalities in both children. Lesions healed within 2.5 months and 1 month of conservative treatment with

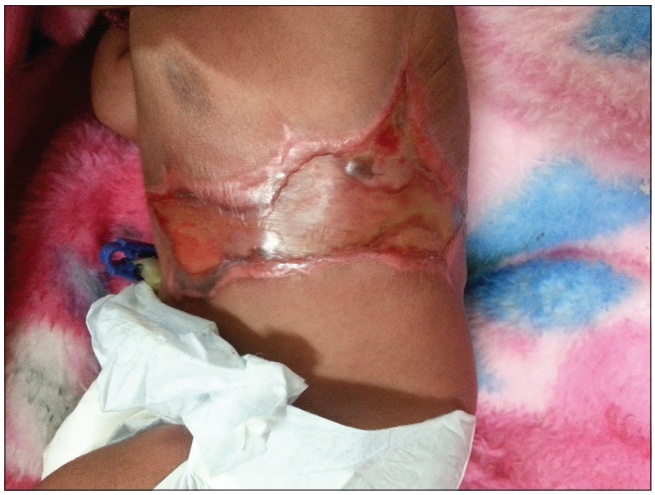

Figure 1a: Absent portion of the skin on the abdomen in a 2-day old male child

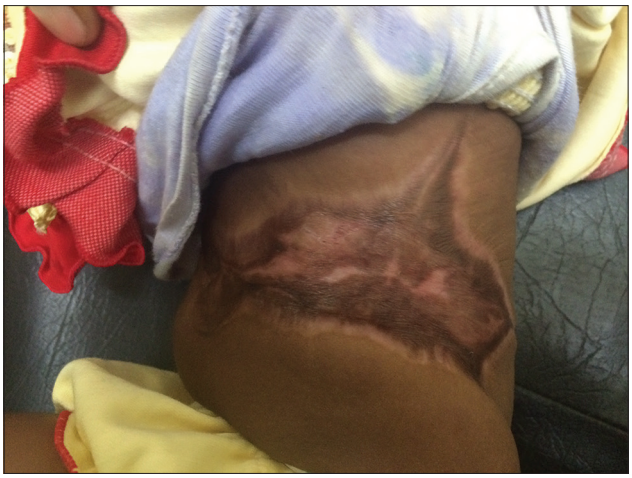

Figure 1b: Complete resolution of lesions after 2.5 months leaving behind a smooth, hypo- to hyperpigmented, hairless, papery scar

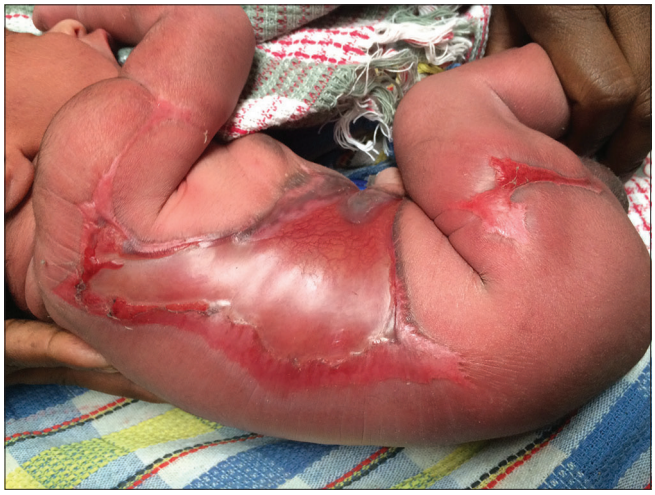

Figure 2a: Typical H-shaped distribution of aplasia cutis congenita in a newborn

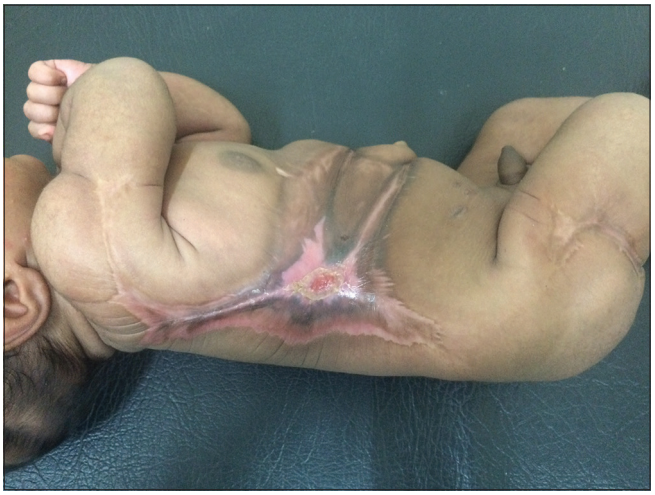

Figure 2b: Complete resolution of lesions after 1 month

topical mupirocin twice daily in case 1 [Figure 1b] and 2 [Figure 2b], respectively.

A diagnosis of aplasia cutis congenita, group 5 without fetus papyraceus was made in both cases.

Frieden classified aplasia cutis congenita into nine groups in 1986.[3]

- Group 1: Aplasia cutis congenita of scalp without multiple anomalies
- Group 2: Aplasia cutis congenita of scalp with limb anomalies
- Group 3: Aplasia cutis congenita of scalp with epidermal and sebaceous nevi
- Group 4: Aplasia cutis congenita with a hair collar overlying deeper embryonic malformations such as meningomyelocele, porencephaly and others
- Group 5: Aplasia cutis congenita associated with fetus papyraceus/placental infarcts
- Group 6: Aplasia cutis congenita associated with epidermolysis bullosa
- Group 7: Aplasia cutis congenita of the extremities without epidermolysis bullosa
- Group 8: Aplasia cutis congenita associated with teratogen exposure, intrauterine infections or drugs
- Group 9: Aplasia cutis congenita associated with malformation syndromes such as Patau syndrome, Goltz syndrome and others.

During a twin pregnancy, the intrauterine death of one fetus occurring at the 12–14th week of gestation may result in the persistence of a dead fetus (fetus

papyraceus) in association with a live and viable twin. Fetus papyraceus causes the release of thrombogenic substances that can cause placental infarction, disseminated intravascular coagulation and cutaneous lesions.[4] In singletons with no history of fetus papyraceus, as seen in our cases, such lesions reflect either *in utero* death of an unrecognized twin/triplet or placental infarction.[2] Typhoid in the mother in the second case might be the cause of truncal aplasia cutis congenita. Although intrauterine typhoid infection has not been implicated in the causation, it has a great impact in causing intrauterine death. *Salmonella typhimurium* proliferates in the infected placenta and causes placental necrosis thus leading to fetal death.[5]

Most lesions heal spontaneously from the margins, leaving behind a smooth, yellowish, hairless, papery scar. Small residual lesions can be treated by excision of the abnormal skin margins followed by primary closure. Composite skin grafts have been used successfully, using allogenic keratinocytes or autologous fibroblasts followed by keratinocytes a week later.[2]

We were unable to find similar previous reports. Truncal aplasia cutis congenita is itself a rare presentation, more so in the absence of history of twin pregnancy/papyraceus fetuses. These cases also draw attention to the possibility of *in utero* death of an unrecognized twin/triplet. Further, the role of other hidden factors or the possibility of intrauterine infection such as typhoid needs to be studied in the causation of truncal aplasia cutis congenita. More evidence is needed to confirm the entity of aplasia cutis congenita, group 5 without fetus papyraceus.

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Conflicts of interest
There are no conflicts of interest.

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