A Rare Case of Type-I Nonsyndromic Aplasia Cutis Congenita

Sir,

With this letter, we would like to engage the attention of the readers toward the condition of Aplasia Cutis in neonates, which if located in the occipital region of the scalp look alike encephalocele. Aplasia Cutis Congenita (ACC) is a rare congenital malformation with worldwide incidence of 0.5–1 per 10,000 births. There is localized congenital absence of the skin of varying severity, ranging from absence of epidermis, dermis, and in some cases, subcutaneous tissues or bone. Around 70% of cases involve the scalp region, and rarely the trunk and limbs. Many etiological factors have been described in the literature - chromosomal abnormalities, intrauterine trauma, teratogen exposure (misoprostol, methimazole, cocaine, methotrexate, angiotensin-converting enzyme inhibitors, benzodiazepines, and valproate), placental thrombotic and vascular events.[1-5]

A thorough work-up of the patient is mandatory to rule out associated malformations. The condition may occur in isolation or as part of a heterogeneous group of syndromes. There may be association with conditions such as cleft lip and palate, trachea-esophageal fistula, heart defects, omphalocele, gastroschisis, polycystic kidney, limb reduction defects, syndactyly, club foot, neural tube defects, hemangioma, mental retardation, seizures, and developmental delay. Frieden classified ACC into nine groups based on the number and the presence/absence of other anomalies. Type I ACC is sporadic or autosomal dominant in inheritance and involves only scalp without multiple anomalies.[1-5] Small defects heal spontaneously, but larger defects require surgical intervention in the form of skin grafts, rotation flaps, free flaps, and tissue expansion. Complications of ACC include infection, hemorrhage, thrombosis, and seizures, which are more likely in ACC with underlying skull bone defect.[1-5]

We managed a term baby boy who was born to fourth gravida euthyroid mother with the past history of one spontaneous abortion during the 13th week of gestation and threatened abortion in the index case. There was neither history of consanguinity, drug or radiation exposure during pregnancy, nor any history suggestive of *in utero* viral exposure. The baby was born vaginally without any instrumentation. At birth, the baby was noted to have a full-thickness oval-shaped skin defect 4 cm × 2 cm over scalp in the occipital region [Figure 1a]. The ulcer was nonindurated without any signs of inflammation in the surrounding skin. The rest of the scalp and skin over the body was normal. There were no other signs of dysmorphism, and the vitals and systemic examination were normal. X-ray of the skull and skeletal survey was normal. Considering the location of the defect, magnetic resonance imaging scan of the head was done, which ruled out underlying bone defect and occipital meningocele. A screening ultrasound of the abdomen, to rule out renal anomalies was normal. Screening echocardiographic evaluation found a small 3 mm atrial septal defect with left to right blood flow. Eye and ear, nose, throat examinations; newborn thyroid screening results and karyotyping were normal.

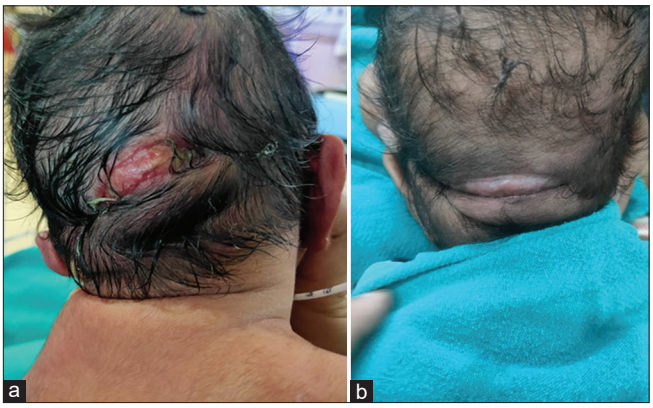

Figure 1: (a and b) Isolated full-thickness aplasia cutis congenita over scalp with signs of complete epithelization over 3 months with conservative management protocol

The skin defect was managed conservatively with daily wound cleaning with povidone–iodine solution and coverage with a sterile dressing. The baby was discharged on day 25th of life with an explanation of prognosis and to continue dressing of the defect at home once the mother was confident of it. The baby was followed till 2 months of age, and the lesion healed completely by then [Figure 1b]. The development milestones achieved were also appropriate to age by the time of last follow-up.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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Submitted: 02-Mar-2023 **Revised:** 26-Jul-2023
Accepted: 04-Dec-2023 **Published:** 22-Dec-2023

Access this article online

Quick Response Code:

Website: www.actamedicainternational.com

DOI: 10.4103/amit.amit_22_23

How to cite this article: Gupta P, Mitra A. A rare case of type-I nonsyndromic aplasia cutis congenita. Acta Med Int 2023;10:160-1.