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Case Report: Non Syndromic Aplasia Cutis Congenita of the Scalp with Bone Defect and an Exposed Sagittal Sinus in Normal Near Term Newborn

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INTRODUCTION

Aplasia cutis congenita (ACC) is a rare and heterogeneous disorder characterized by congenital absence of skin. The scalp is the most commonly affected site and lesions may overlie deeper ectodermal abnormalities (1), It is a focal deficiency of cutaneous tissues of arying severity, ranging from an absence of skin through to full thickness defects involving deeper elements such as bone and dura. Lesions of the scalp can be associated with complications including infection, hemorrhage, thrombosis, and seizures. It rarely occurs on the trunk and limbs, and can occur in isolation or as part of a heterogeneous group of syndromes(1)

The incidence of aplasia cutis is 0.5/10,000 to 1/10,000 newborns; the ratio female/male newborns is around 7:5.

Diagnosis of ACC is clinical with appearance variability. Physical examinations may show ulcerations or erosions of the skin, which may extend to a deeper tissue, such as muscle or bone,particularly the vertex region. Bone abnormalities in ACC are found in approximately 15–20% of cases,ACC classification based on Frieden includes nine subtypes associated with abnormalities, inheritance patterns, and affected body areas, as shown in Table 1 (2, 3),

Aplasia cuties congenita classification

Type	Description
1	Scalp ACC without multiple anomalies
2	Scalp ACC with associated limb abnormalities
3	Scalp ACC with associated epidermal and organoid naevi
4	ACC overlying embryological malformations
5	ACC with associated fetus papyraceous or placental infarcts
6	ACC localized to extremities without blistering
7	ACC caused by specific teratogens
8	ACC associated with a malformation syndrome
9	Unclassified

The exact mechanism is still not completely understood, although many etiological factors have been incriminated in recent years: [4)]

- chromosomal abnormalities,
- traumatic mechanism,
- amniotic defects,
- intrauterine problems,

- thrombotic events, vascular alterations.[5]
- teratogens used in pregnancy: misoprostol, cocain, methotrexate, angiotensin-converting enzyme inhibitors, methimasol, benzodiazepines, valproic acid

In this article, we report a case of aplasia cutis congenita of the scalp with dura and bone defect and an exposed sagittal sinus the defect measuring about 9.7 x 10.5 in a normal nearterm newborn AGA,with no dysmorphic features ,no limb anomalies away of amputated tips of the toes of both feet with normal karyotyping

2. Case description

The patient was a 1-day-old Saudi near term boy born to a 31-year-old lady at 36 weeks of gestation via a cesarean section due to previous cesarean section, Maternal history revealed that the mother was gravida 2 para 1 with history of pregnancy induced hypertension on regular intake of labetalol for control of hypertension no known chronic medical illness. She was doing her antenatal care regularly. She took supplement medication during pregnancy, including folic acid, iron, and calcium, with good adherence. There was no history of gestational diabetes or radiation exposure. Her previous pregnancy was a cesarean section of term boy from an ex-husband, in good condition with no chronic illness. With regard to family history, the parents are not consanguineous.

The APGAR scores were 6,7, and 8 at 1, 5 and 10 min at the delivery room, respectively. He was admitted to a neonatal intensive care unit (NICU) because of aplasia cutis congenital of the scalp extending posteriorly involving whole vault of the skull and extend anteriorly with full exposure of sagittal sinus and brain due to absence of whole scalp thickness and underlying bone and mild respiratory distress, which improved rapidly on nasal cannula oxygen , physical examination revealed a newborn boy vital signs were within acceptable values, and he had pinkish skin color and with no pallor, jaundice, or cyanosis ,no hyopmelanocytic macules or skin rash with normal skin across the body . His growth parameters based on age and sex were in the 25th percentile for weight, length, and head circumference. He had no dysmorphic facial features,He had an area of loss of scalp skin and skull bone with visible brain tissue and an exposed sagittal sinus that measured 9.7 by 10.5 cm in size, as shown in Figure 1., upper limbs were normal while lower limb examination showed amputated tips of the toes in both feet suspected amniotic band disruption Lung auscultation revealed bilateral vesicular breathing with equal air entry and no added sounds. Cardiac auscultation revealed nomurmur, He had a soft and lax abdomen without tenderness or hepatosplenomegaly.

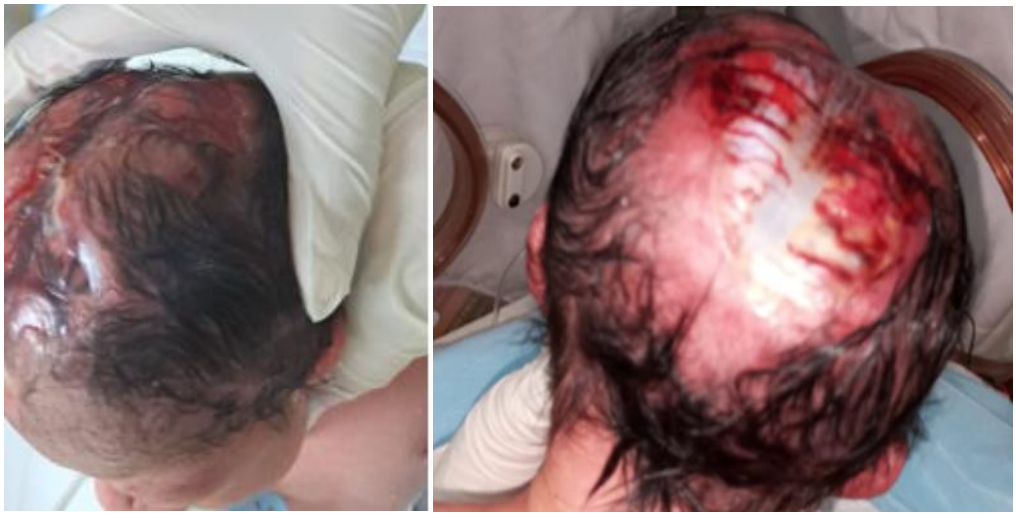
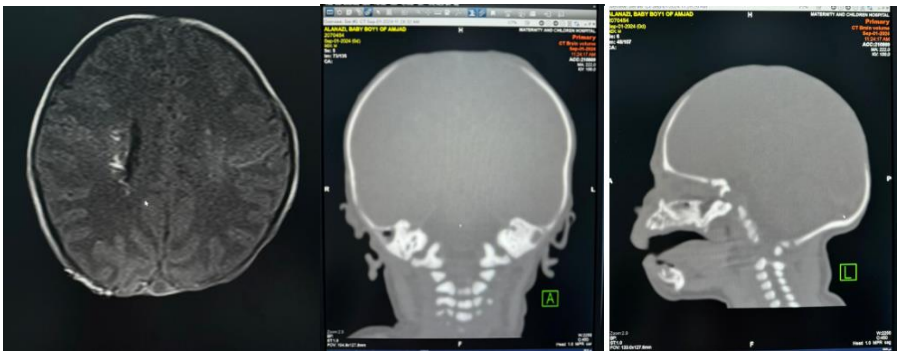


Figure (1). aplasia cutis congenita of the scalp

Laboratory investigations at the age of 24 hours included basic workups within the normal range for her age. The patient's blood group was O-positive, and A liver function test showed total bilirubin of 60 mmol/l, direct bilirubin of 5.2 mmol/l, SGOT of 25 U/L, SGPT of 10 U/L, total protein of 5.1 g/dl, and albumin of 3.1 g/dl. Virology screening, including the TORCH test (toxoplasma, rubella, cytomegalovirus, and herpes simplex virus), was negative. Hepatitis B surface antigen and antibody

were non-reactive and the core antibody was negative. The blood culture did not show bacterial growth. chromosomal analysis showed a normalmale karyotype
Radiology investigations included head magnetic resonance imaging (MRI) and (CT) revealed absence of scalp and calvarium at the parietal region with no herniation visualized, also sub ependymal periventricular calcified nodules as shown in Figure 2 a ,b &c



(a)Axial T1 sequencewith right sub ependymal calcifications
(b)Coronal & sagittal CT brain bone window

3D skull reformat (fig 2)
Figure (2) Selected image of axial CT brain showing

(c)Sagittal CT brain bone window
Selected images of CT brain in bone window with 3D reconstruction showing large area of absent calvarium involving bilateral frontoparaital bones(fig 2)

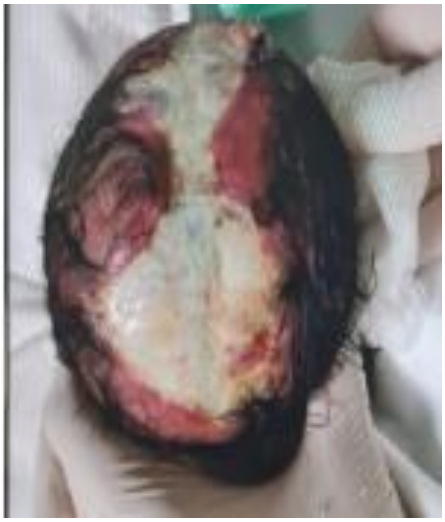
Which initially raise the suspension of tuberous sclerosis versus TORCH infection
Echocardiography was done and revealed no any intracardiac lesions or tumers only small ASD & tiny closing PDA
Abdomen and pelvis ultrasonography were normal with no any vascular renal problems or cystic kidney diseases
Baby started on ampicillin &cefotaxime meningeticdose. urgent neurosurgery and plastic surgery consultation was done and baby managed by multidisplinary team
As baby was clinically stable with no CSF leak, the case managed conservative without intervention and spontaneous epithelializationstarted (fig 3)

3. Discussion

Aplasia cutis congenita (ACC) is a rare and heterogeneous disorder characterized by congenital absence of skin. The scalp is the most commonly affected site and lesions may overlie deeper ectodermal abnormalities. The exact etiology is still unknown. Chromosomal analysis is recommended for any newborn with ACC, with high consideration if there are other congenital anomalies. (1)

Skull or dura defect in ACC results in brain and sagittal sinus exposure. Such exposure increases the risk of hemorrhage, infection, and sagittal sinus thrombosis. The reported fatality of ACC is 20–50% (6). The preoperative evaluation includes history, physical examination, and radiological investigation

by three-dimensional facial bone computed tomography (CT) and magnetic resonance imaging (MRI) of the brain (6). The guiding prioritization criterion and the choice of intervention remain controversial and vague. However, it is important for neurosurgeons to decide promptly whether to perform early surgical intervention or to proceed with conservative care (6). For example, small lesions with noninjured dura and small bone defects that do not overlie the superior sagittal sinus can be managed conservatively by a gauze dressing with saline drips, topical antibiotic ointment, povidone-iodine, and silver sulfadiazine, which hopefully will heal gradually with re-epithelization FIGUAR (3)



Figour 3 showed re-epithelization under consarvative manegement

The goal of such management is to ensure and maintain a moist healing environment to avoid Escher formation and minimize bleeding risk and CSF leakage. Meanwhile, large defects with full thickness involvement of scalp and bone require more time for complete closure as they require a longer procedure (7). However, large scalp and skull defects can be managed conservatively with complete healing, as has been reported (6). Surgical intervention indication
includes enlarged vein exposure, associated dura defect, and brain exposure. Such surgical options include split-thickness or full-thickness skin grafts, scalp rotation flaps, pericranial flaps, split rib grafts with latissimus dorsi muscle flaps, and tissue expansion (7).
Surgical management complications include hemorrhage, graft or flap loss, infection, donor site morbidity, and anesthesia-related complications (1, 6). However, conservative management may develop complications, such as massive hemorrhage, cerebrospinal fluid leak, central nervous system infection, and sepsis. In contrast, in cases of dural defects, the patient may have herniation of the brain with parenchymal injury and cerebral necrosis (6, 7). Additionally, if ACC occurs due to a genetic disorder, it will further burden the surgical complication if there are other comorbidities. (8)
In conclusion, the history and physical examination of ACC can guide healthcare providers to the underlying pathophysiology and further evaluations and treatments can be performed if needed. Family counseling is important to provide understandable family values and goals, as well as to achieve the expected outcomes and disease trajectories (4). The case emphasizes the importance of ACC management, a comprehensive approach that involves the clinical status of the patient to determine the best surgical and non-surgical management.

The patient was managed with a moist gauze dressing, topical antibiotic ointment, and povidone-iodine. A multidisciplinary team meeting was held that involved a neurosurgeon, a neonatologist, and pediatric genetics consultants

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