CASE REPORT

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Aplasia Cutis Congenita of the Scalp with Bone Defect and Exposed Sagittal Sinus in Trisomy 13 Newborn – a Case Report

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ABSTRACT

Background: Aplasia cutis congenita is a heterogeneous disorders group with a rare reported incident of 0.5 to 1 in 10,000 births. ACC can be associated with physical defects or syndrome that may help in diagnosis, prognosis and further evaluation of the patient. Trisomy 13 is one of the most common fetal life limiting diagnosis which is associated with ACC of membranous type scalp. **Objective:** In this article, we report cases of aplasia cutis congenita of the scalp with dura and bone defect and exposed sagittal sinus in newborn diagnosed to have trisomy 13. It emphasizes the importance of ACC associated syndrome which is having high mortality prior to surgical intervention. **Case presentation:** The patient was born at 35 weeks of gestation. Her physical examination revealed a newborn girl with dysmorphic facial features including widely separated eyes, downward slanting of the palpebral fissure, microphthalmia, retrognathia, and low seat ears. She had area of loss of scalp skin and skull bone with seen brain tissue and sagittal sinus were exposed that was measure 6 by 5 cm in size. Additionally, she had a clenched fist and overlapping fingers and rocker bottom feet. Laboratory investigations include basic labs and the TORCH screen was negative. On the 9th day of life, a chromosomal analysis showed a female karyotype with three copies of chromosome number 13 in all 20 metaphase cells counts. **Conclusion:** The patient was managed conservatively. However, a multidisciplinary team agreed on do not resuscitate with no further surgical intervention as survival rate of trisomy 13 is poor.

Keywords: Aplasia cutis congenita, trisomy 13, Patau syndrome, surgical managmentm non-surgical managment.

1. BACKGROUND

Aplasia cutis congenita (ACC) is a description of focal, localized congenital absence or defect in the skin with heterogeneous clinical presentations (1, 2). ACC is rare and reported incidence is 0.5 to 1 in 10,000 births (1). The pathogenesis of ACC is unknown and it can be divided into two main pathways: first is skin layers development disruption or failure that include the epidermis, dermis and subcutaneous fat, and secondly is a skin destruction in-utero that was developed normally (1).

Diagnosis of ACC is clinical with appearance variability. Physical exams may show ulcerations or erosions of the skin which may extend to a deeper tissue such as the muscle or bone. Also, it may appear as an atrophic scar. Around 86% of Aplasia cutis congenita cases involve the scalp which is mainly the vertex. Bone abnormalities in ACC is approximately 15 to 20% of cases as the remaining majority involve the scalp (1, 2). ACC can be associated with physical defects or syndrome in which it is important to identify the various clinical subtypes of ACC that will help in diagnosis, prognosis and further evaluation of the patient (1). ACC classifications based on Frieden include nine subtypes on its associated abnormalities, inheritance pattern and affected body area as shown in Figure 1 (1, 2). While Evers classify ACC into four group based on (1) chromosomal changes, (2) monogenic group that include autosomal dominant or recessive and X-linked genetic mutations, (3) teratogenic/exogenous causes, and (4) unknown group that include ACC with 2 or more congenital defects or single congenital defect with uncertain cause (1).

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 papyraceus or placental infarcts
6	ACC localized to extremities without blistering
7	ACC caused by specific teratogens
8	ACC associated with malformation syndrome
9	Unclassified

Table 1. ACC classifications

2. OBJECTIVE

In this article, we report cases of aplasia cutis congenita of the scalp with dura and bone defect and exposed sagittal sinus in newborn diagnosed to have trisomy 13. It emphasizes the importance of ACC associated syndrome which is having high mortality prior to surgical intervention.

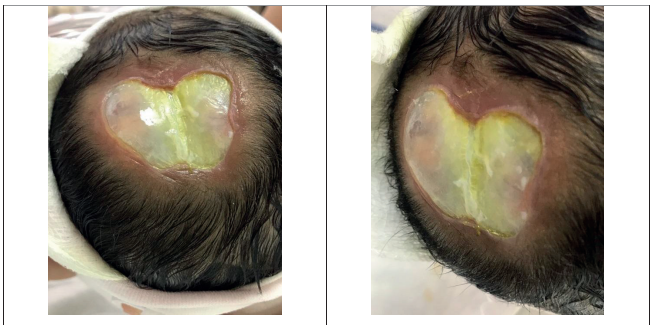
3. CASE PRESENTATION

The patient was a 23 day old Saudi girl who was born to a 41 years old lady at 35 weeks of gestation via cesarean sections due to fetal distress. Maternal history revealed the mother was gravida 5 para 6 with no known chronic medical illness. She started her follow up in the private sector in which first ultrasound showed a single viable fetus but unfortunately an anomaly scan was not done. She was taking supplement medication during pregnancy once she tested positive on a pregnancy test as used to take folic acid, iron and calcium with no good compliance on taking them at times. There was no history of gestational diabetes or pregnancy induced hypertension as she was screened during pregnancy follow-up. No history of radiation exposure. Her previous 4 pregnancy concluded with first normal vigainal delivery and at 2nd was cesarean section due to abruption placenta and 3rd was cesarean section of twins delivery and the fourth was cesarean section due to previous cesarean section. However, all children were full term and in good condition with no chronic medical illness. In regards to family history, the parents are not consanguineous. There is one maternal aunt with trisomy 21.

At the delivery room, the APGAR score was 5 and 7 at 1 and 5 minutes respectively. She was admitted to NICU due to respiratory distress which started on CPAP initially then required intubation on the 4th day of life. On the 5th day of life she was transferred to our hospital due to limited other service and financial issues. Upon admission to our hospital, her physical examination revealed a newborn girl under radiant-warmer and intubated with connection to mechanical ventilations. Her vital signs were within acceptable values and she was pinkish skin color and not pale, jaundiced, or cyanosed. Her growth parameters based on age and sex were in the 10th percentile for weight, length, and head circumference. She had dysmorphic facial features including widely separated eyes, downward slanting of the palpebral fissure, microphthalmia, retrognathia, and low seat ears. She had area of loss of scalp skin and skull bone with seen brain tissue and sagittal sinus were exposed that was measure 6*5 cm in size as shown in Figure 3. Additionally, she had a clenched fist and overlapping fingers and rocker bottom feet. Lung auscultation revealed vesicular breathing with equal air entry bilaterally and no added sounds. Precordium auscultation revealed medium pitched high grade continuous murmur heard best at the pulmonary position

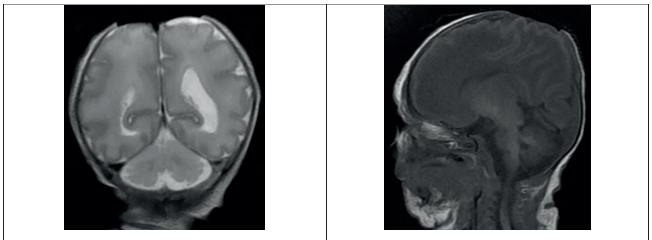
Chromosomal	Patau syndrome, Pallister-Killian syndrome, Wolf–Hirschhorn syndrome
Autosomal Dominant	Adams-Oliver syndrome, Autosomal dominant ACC, Ectrodactyly-ectodermal dysplasia-clefting syndrome, Ectodermal dysplasia, Scalp-Ear-Nipple syndrome
Autosomal	Recessive Autosomal recessive ACC, Johanson-Blizzard syndrome, Setleis syndrome, Ectodermal dysplasia-clefting syndrome, Epidermolysis bullosa
X-Linked	Goltz-Gorlin syndrome (focal dermal hypoplasia), MIDAS (Microphthalmia, Dermal Aplasia and Sclerocornea) syndrome
Teratological/ Exogenous	Alcohol, cocaine, marijuana, methimazole, misoprostol, Congenital infections (herpes simplex, rubella, varicella), Amniotic band disruption complex

Table 2. ACC associated congenital anomalies



Newborn head showing 6 by 5 cm full-thickness defect of the scalp , skull and dura. You can notice arachnoid layer covering the brain with noted brain tissue. Also notice exposed superior sagittal sinus covered with a layer of granulation.

Figure 1. Patient skin defect



Selective images of the MRI coronal T2 and sagittal T1 weighted images show absence of scalp and calvarium at the parietal region with no herniation .

Figure 2. Patient head MRI

with a harsh machinelike quality that often radiated to the left clavicle. She had a soft and lax abdomen without tenderness or hepatosplenomegaly.

Laboratory investigations at the age of 5 days included complete blood count (CBC) which revealed a hemoglobin of 12.1 g/dl, haematocrit of 35.8 %, MCV of 113 fL, MCH of 38.2 pg, MCHC of 33.9 g/dl, RDW of 16.9 %, platelet of 121 k/ul, WBC of 23.6 k/ul in which neutrophils were 74 % lymphocyte of 11% and monocyte of 15 %. Patient blood group is O positive and the direct coombs test (DCT) was negative. Liver function test showed total bilirubin of 13.7 mg/dl, direct bilirubin of 1.1 mg/dl, SGOT of 75 U/L, SGPT of 14 U/L, lactate dehydrogenase (LDH) of 889 U/L, GGTP of 118 U/L, total protein 4.1 g/dl and albumin of 2.7 g/dl. Renal function test showed a BUN of 13 mg/d, creatinine of 0.82 mg/dl, sodium of 138 mEq/L, potassium of 3.5 mEq/L, CO2 of 18 mEq/L, Cl 103mEq/L, anion gap 17. C-reactive protein level was less than 0.1 mg/dL. Virology screens including Toxoplasma IgG and IgM was negative, Rubella IgG was positive and IgM was negative, Cytomegalovirus (CMV) Antibody IgG was positive and IgM was negative, Herpes Simplex Virus IgG was positive and IgM was negative, Hepatitis B surface antigen and antibody were non reactive and core antibody was negative. Blood culture did not show bacterial growth. On the 9th day of life, a chromosomal analysis showed a female karyotype with three copies of chromosome number 13 in all 20 metaphase cells counts. Radiology investigations include head magnetic resonance imaging (MRI) revealed absence of scalp and calvarium at the parietal region with no herniation is visualized as shown in Figure 4.

The patient was managed with moist gauze dressing, topical antibiotics ointment, and povidone-iodine. A multidisciplinary team meeting was held that involved neurosurgeon, neonatologist, and pediatric genetics consultants who agreed on do not resuscitate (DNR) with no further surgical intervention as survival rate of trisomy 13 is poor along with associated skull bone defect.

4. DISCUSSION

In 1826, Campell was the first who report aplasia cutis congenita. Chromosomal analysis is recommended to be taken in any newborn with scalp ACC with high consideration if there are other congenital anomalies as genetic causes of ACC as shown in Figure 2 (6). Such associated anomalies is Trisomy 13 which is one of the most common fetal life limiting diagnosis with a prevalence of 1.68 per 10,000 births (3). Approximately 10 days is the median survival days for live births with trisomy 13 (4). Anomalies associated with trisomy 13 include 34% with major cardiac anomalies, 25% with oro-facial clefts, and 11% with nervous system anomalies (4). ACC of scalp type is a common association with trisomy 13 but bone associated is rare (1).

Skull or dura defect in ACC result in brain and sagittal sinus exposure. Such exposure increases the risk of hemorrhage, infections, and sagittal sinus thrombosis. The reported fatality of ACC is 20 to 50% (5). In which preoperative evaluation include history, physical examination and radiological investigation as 3-Dimensional facial bone computed tomography (CT) and magnetic resonance imaging (MRI) of the brain (5). Guiding prioritization criteria and choice of interventions 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 (5). Such as small lesion with non-injured dura and small bone defect that is not overlie the superior sagittal sinus it can be managed conservatively that include gauze dressing with saline drips, topical antibiotics ointment, povidone-iodine, and silver sulfadiazine which hopefully will heal gradually with re-epithelization.

The goal of such management is to ensure and maintain a moist healing environment to avoid eschar formation and minimize bleeding risk. Meanwhile in large defects with full thickness involvement of scalp and bone it required longer months for complete closure as it takes longer procedure (7). However, large scalp and skull defects can be managed conservatively as it has been reported with complete healing (5). Surgical intervention indication includes enlarged vein exposure, associated dure defect and brain exposure. Such surgical options include split-thickness or full-thickness skin graft, scalp rotation flaps, pericranial flaps, split rib graft with latissimus dorsi muscle flap and tissue expansion (7).

5. CONCLUSION

In conclusion, history and physical examination of ACC can guide health care providers to the underlying pathophysiology and need further evaluations and treatments if needed. Family counseling is important to provide understandable values and goals of the family, as well as the expected outcomes and disease trajectory (3). The case emphasizes the importance of ACC management, a comprehensive approach that involves clinical status of the patient that will determine the best surgical and nonsurgical management.

- **Patient Consent Form:** Consent was obtained from the patient parents and the authors for publication of this case report.
- **Author's contribution:** AAS and FR reviewed the literature. AAS and FR wrote the introduction and discussion. RN and Ab wrote the case. AAS and FR wrote the first draft of the manuscript. RN and AMMAR read and approved the final version of the manuscript.
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