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Aplasia Cuties Congenita: A Case Study and Review of Literature Updates

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Abstract | **Case Report**

Cordon, in the year 1767, was the first person to describe the extremely rare form of abnormality known as congenital aplasia of the skin. On the scalp, limbs, or abdomen, it manifests as a single, isolated lesion that affects multiple layers of skin and, on occasion, the bone. This is the most common form in which it presents itself. There has been some speculation that the condition could have a genetic component, a component related to the environment, or a component related to an exogenous factor. As of the end of 2013, there had been a total of approximately 500 cases reported from all over the world. The aim of the present study was to describe a rare case of ACC. An infant was diagnosed with ACC and a scalp lesion. The lesion was followed and described over one year.

Keywords: Aplasia cuties congenita, scalp, lesion, genetics, skin.

INTRODUCTION

Aplasia cuties congenita (ACC) is a form of congenital anomaly that affects a very small percentage of people and is characterized by the complete or partial lack of a patch of skin and, in some instances, bone at birth (Lewis *et al.*, 2017). Even though the illness typically begins on the scalp, it has the potential to spread to other parts of the body, including the face, the torso, and the limbs (Tempark and Shwayder, 2012; Duan *et al.*, 2015). Congenital aplasia of the skin is a form of abnormality that is extremely rare and was first described in 1767 by Cordon. On the scalp, limbs, or abdomen, it manifests as a single, isolated lesion that affects multiple layers of skin and, on occasion, the bone (Mukhtar- Yola *et al.*, 2020).

It is generally accepted that the aetiology of ACC is due to the interaction of a number of different causal variables. Some of the hypotheses that have been proposed in connection with its development include incomplete closure of the neural tube and embryonic fusion lines; vascular insufficiency to the skin as a result of placental insufficiency; intrauterine infections; teratogenic agents; amniotic bands; and chromosomal abnormalities (Frieden, 1986; Caksen and Kurtoglu, 2002). This illness has been characterized by nine distinct clinical subtypes that have been recognized. These subtypes can be separated from one another based on the location and pattern of skin absence, the anomalies that accompany the condition, and the manner of inheritance (Frieden, 1986; Tempark and Shwayder, 2012).

There has been some speculation that the condition could have a genetic component, a component related to the environment, or a component related to an exogenous factor (AlShehri *et al.*, 2016). As of the end of 2013, there had been a total of approximately 500 cases reported from all over the world. A child born into a consanguineous family with associated cardiac, skin, and nail anomalies (likely Adams Oliver syndrome) was found to have one case of Aplasia Cutis Congenita (ACC), while the other case was found as an isolated scalp lesion. In both instances, birth examinations revealed problems with the patient's scalp and skeleton. Both were significant flaws that needed to be handled cautiously, which required a group of people with expertise across a wide range of fields (Mukhtar-Yola *et al.*, 2020).

ACC an extremely uncommon form of congenital abnormality that impacts the various layers of the skin. Solitary lesions affecting the midline over the vertex of the skull and, less commonly, the periosteum and bone that lie underneath the skin are the most common manifestations of this condition (Bajpai and Pal, 2003). Additionally, it has been reported in other locations, including the limbs, the abdomen, and the chest (O'Neill *et al.*, 2010). Congenital defect of the skull and scalp, congenital ulcer of the newborn, and

congenital scalp defect are some of the other names that have been given to ACC by the National Organization for Rare Disorders (NORD, 2020). However, it does appear to be multifactorial in some cases, as both genetics and environmental as well as exogenous causes have been implicated (Burkhead *et al.*, 2009; AlShehri *et al.*, 2016). Neither the etiology nor the pathophysiology of ACC is well understood (Mukhtar-Yola *et al.*, 2020). In addition to sporadic cases, there have been reports of cases that exhibit characteristics of both autosomal dominant and autosomal recessive inheritance (Lei *et al.*, 2019). The pathophysiology model that has received the most widespread acceptance describing the tension that prevents the skin from properly approximating during the development of the fetus. An inadequate blood supply to the skin, fetal and placental ischemia, intrauterine infections, and an inability to successfully close the neural tube are some of the potential causes of this condition (Blionas *et al.*, 2017).

When it comes to newborns, the majority of the time, the diagnosis is established during the first examination of the child. The treatment for it varies depending on the size of the defect, where it is located, and the degree to which it involves the structures that are adjacent to it. These factors are all taken into consideration. In order to lessen the likelihood of complications like sagittal sinus hemorrhage, infection, fluid and electrolyte imbalances, or thermal imbalances, treatment ought to get underway as quickly as is humanly possible (Maillet-Declerck *et al.*, 2013).

CASE PRESENTATION

Female newborn baby born on 6/Jun/2021, product of CIS due to a previous CIS in Al-Basheer hospital for a healthy mother and father and unremarkable prenatal history. Term, birth body weight =3kg. The baby was admitted to the Neonatal Intensive Care Unit (NICU) for investigation for the scalp defect that found at birth. Physical examination showed that the baby lacked the skin on the scalp vertex area as well-demarcated circular ulcer about 2×2 cm, which raised up the potential diagnosis of ACC (figure 1).

Figure 1: The status of the lesion at birth

The baby was discharged home from NICU after dressing care and empirical IV ABX till cultures of blood and site of defect were negative with instructions for defect care and follow up in clinic.

At age of 2 months, defects were healed causing alopecia (figure 2).

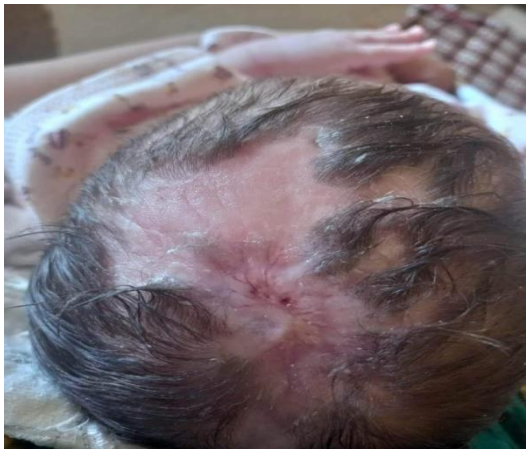

Figure 2: Healing improvement at 2 months

Because there is risk of associated neurological defect brain, CT-scan was carried out at the age of 2 months and showed the following findings: area of encephalomalacia was seen in the right MA territory (fronto -parietal and temporal region) with foci of calcification with evacuee dilatation of ipsilateral ventricle. No evidence of cortical calcification or calvarias thickening and advised to do MRI later. The follow up in clinic in first year of life showed some of developmental delay and slowed down of head circumference (figure 3).

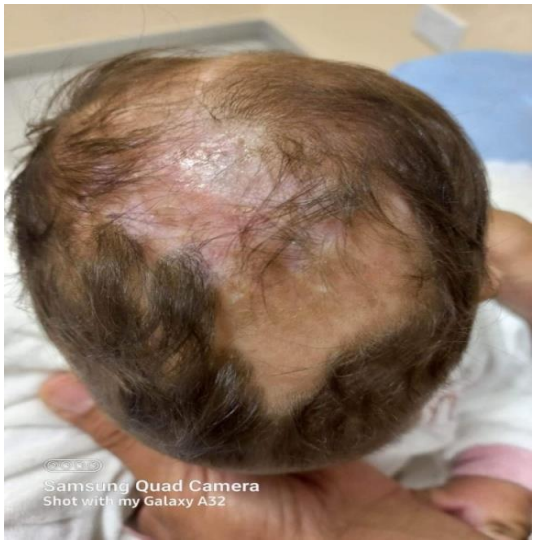

Figure 3: improvement of the lesion at 6 months

On follow up at age of 1 year, patient was unable to stand. The findings of MRI showed extensive area of encephalomalacia involving the right frontal temporoparietal lobes with adjacent gliosis, along with

ex vacuo dilatation of the right lateral ventricle. Small sized right basal ganglia with Wallerian degeneration of the right cerebral peduncle. Confluent hyperintense T2 weighted and FLAIR sequences of the periventricular and subcortical white matter bilaterally, to be correlated for patient's clinical data. Dysplastic corpus callosum was also observed. The findings also showed no focal brain lesion. However, minimal shift of midline structures to the right existed. There were no acute brain insults or hemorrhage. The patient was sent for physiotherapy and was recommended to subject for cosmetic surgery for scalp skin graft in future.

DISCUSSION

Because there are no particular histological alterations associated with the condition (Bajpai and Pal, 2003), the diagnosis of aplasia cutis congenita is predominately made through clinical examination. One case of ACC with birth defects in the scalp has been reported in this study. The case occurred in a child who had a syndromic condition. The infant had skin defects that appeared as large sheets that were asymmetrical in shape and involved hypoplasia of the subcutaneous tissues in addition to the bones. Frieden and Schierz discovered that 70% to 90% of lesions are localized to the vertex of the scalp, presenting as solitary or multiple lesions that are non-inflammatory, well demarcated, sheet-like, or appear as patches with irregular shapes, size, and involving variable skin depths (Frieden, 1986; Schierz *et al.*, 2020). It is possible to have a single lesion with these characteristics, or to have multiple lesions with the same characteristics. Others have mentioned that the shape of the lesion could be circular, oval, linear, or stellate (Mukhtar-Yola *et al.*, 2020). The close proximity of scalp aplasia cutis congenita to the scalp hair whorl, which is believed to be the point of maximum tensile force during rapid brain growth, has led to the hypothesis that tension-induced disruption of the overlying skin occurs between the ages of 10-15 weeks of gestation. This is the time period during which hair direction and patterning as well as rapid brain growth take place. In addition to this, this could also explain why there is a higher incidence of aplasia cutis congenita on the vertex of the scalp (Mukhtar-Yola *et al.*, 2020). At the time of birth, the lesions may already be healed with scarring, or they may continue to be superficially eroded or deeply ulcerated, occasionally involving the dura or the meninges (Mukhtar-Yola *et al.*, 2020).

The findings of MRI showed no focal brain lesion. However, minimal shift of midline structures to the right existed. There were no acute brain insults or hemorrhage. Similar findings were reported by case studies reported by Mukhtar-Yola *et al.*, (2020).

CONCLUSIONS

When it does happen, ACC poses a number of difficult challenges to clinicians in pediatric departments. This is especially true when the defects are large or when it is associated with a number of other congenital malformations.

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