Non-surgical management of severe aplasia cutis congenital accompanied by a bone defect – a case study

Monica Surdu1,2, Cristina-Ioana Geanta2, Irina Franciuc1,2, Georgia Ioana Olginda2, Traian Virgiliu Surdu1,3

1“Ovidius” University, Faculty of Medicine, Constanta, Romania
2Neonatology Department, County Clinical Emergency Hospital of Constanta, Romania
3Techirghiol Balnear & Rehabilitation Sanatorium, Constanta, Romania

ABSTRACT

Background and objectives. Aplasia cutis congenita (ACC) is a rare developmental disorder that is not fully understood. It is part of a diverse group of conditions characterized by the congenital absence of the epidermis, dermis, and occasionally subcutaneous tissues or bone, often affecting the crown of the scalp. It frequently presents as an isolated condition but can also be associated with a syndrome. Among patients, scalp involvement is common; however, large scalp defects with skull involvement are uncommon.

Materials and methods. We report the case of a newborn delivered at full term by cesarean section, 3200g, in the Mangalia Municipal Hospital, later transferred to the Constanta Neonatology Department, Neonatal Intensive Care department, for 3 injuries with lack of substance, both in tegument and bone, of different sizes, at the level of the vertex and the occiput.

Results. Given the fragile age, a conservative management approach was chosen. The wound was meticulously cleaned daily using sterile water, and a self-adhesive absorbent dressing with hydrocolloid was applied. This dressing promoted granulation and epithelialization, adjusting dynamically to the lesion size. The newborn remained hospitalized for 35 days until the closure of the most extensive skin lesion at the vertex.

Conclusions. Aplasia cutis congenita can be managed using various treatment approaches, but a consensus on the optimal strategy remains elusive. While conservative treatment has been proposed, several authors have highlighted its limitations. The choice between conservative and surgical management should be personalized, considering factors such as lesion size, location, underlying etiology, and related abnormalities.

Keywords: aplasia cutis congenita, scalp, skin defect, surgical management, non-surgical management, cranial reconstruction, scalp reconstruction, newborn, preoperative stabilization

INTRODUCTION

Aplasia cutis congenita encompasses a diverse range of disorders initially documented by Cordon in 1767. The lack of epidermis or dermis defines the condition, and sometimes subcutaneous tissues, or even bone tissue, are associated. These anomalies can occur in various locations on the body. The scalp is the most frequent location (up to 70% of cases), and any skin area may be affected. ACC is uncommon, with a reported incidence of 0.5 to 1 in 10,000 births. Although the underlying pathogenesis of ACC remains elusive, it can be classified into two main pathways: 1. disruption or failure of skin layer development: this pathway involves the absence of epidermis, dermis, and subcutaneous fat due to developmental anomalies; 2. in-utero skin destruction: in some cases, skin destruction occurs during fetal development despite normal initial development. Multiple factors have been suggested as possible triggers for ACC, such as placental infarctions, genetic influences, exposure to teratogenic agents, intrauterine infections, trauma, compromised blood flow, amnion rupture, adhesions between the amniotic
membrane and fetal skin, amniotic band sequence, ectodermal dysplasia, incomplete neural tube closure, and maternal use of drugs during labor [1-3]. The evaluation of ACC relies on clinical assessment due to its diverse presentation. Skin lesions, sometimes penetrating deeper tissues like muscle or bone, are noticed at the physical examination. Furthermore, ACC lesions can appear as atrophic scars. Roughly 86% of ACC cases affect the scalp, with the vertex being the primary site of involvement. Bone anomalies are present in approximately 15 to 20% of cases, with the majority still related to scalp involvement. Aplasia cutis congenita may be correlated with physical abnormalities or syndromes.

Recognizing the different clinical subtypes of ACC is essential for a precise diagnosis, prognosis, and subsequent patient assessment. Frieden’s classification system comprises nine subtypes based on associated abnormalities, inheritance patterns, and affected body areas - Table 1. Evers’ classification divides ACC into four groups: chromosomal changes, a monogenic group, which includes autosomal dominant or recessive and X-linked genetic mutations; teratogenic/exogenous causes; and an unknown group, which encompasses ACC with two or more congenital defects or a single congenital defect with an uncertain cause. The treatment for ACC can differ depending on the infant’s condition, and conservative management is typically the preferred approach. Although there have been instances of surgical intervention for ACC, the utilization of the scalp as a donor site is seldom documented [3-5].

**TABLE 1.** Classification of the ACC by Frieden, based on their number, location, and associated deformities

<table>
<thead>
<tr>
<th>Characteristics of Types</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
</tr>
<tr>
<td>2</td>
</tr>
<tr>
<td>3</td>
</tr>
<tr>
<td>4</td>
</tr>
<tr>
<td>5</td>
</tr>
<tr>
<td>6</td>
</tr>
<tr>
<td>7</td>
</tr>
<tr>
<td>8</td>
</tr>
<tr>
<td>9</td>
</tr>
</tbody>
</table>

At birth, the neonate exhibited a bone and tegumentary defect at the vertex level, along with two other small lesions (Figure 1). Physical examination revealed no other significant findings. Notably, the newborn’s father and paternal grandmother also had a small skin defect (approximately 2 × 2 cm) at birth, which did not require long-term treatment as it did not impact the underlying bone tissue. Ultrasounds during pregnancy were not performed.

**FIGURE 1.** ACC at the time of birth

**RESULTS**

The integumentary and bone defects were confirmed through cerebral ultrasound and cerebral MRI. The MRI scan revealed and confirmed lesions characterized by a lack of substance with distinct and regular edges. These lesions were observed at the level of the paramedian parietal bones bilaterally, with a maximum axial diameter of 42 mm, as well as at the vertex, with a maximum axial dia-

**MATERIALS AND METHODS**

We describe a case study involving a neonate delivered at 38 weeks of gestation, weighing 3200 grams. The mother, aged 28, had an uncomplicated pregnancy devoid of any history of drug usage, trauma, or infectious ailments. The parents were not con-sanguineous. The infant was delivered via cesarean section.
Multiple interdisciplinary consultations were conducted to determine the best course of action for the neonate. Given the fragile age, a conservative management approach was chosen. The wound was meticulously cleaned daily using sterile water, and a self-adhesive absorbent dressing with hydrocolloid was applied. This dressing promoted granulation and epithelialization, adjusting dynamically to the lesion size (Figure 3). The newborn remained hospitalized for 35 days until the tegumentary closure of the most extensive skin lesion at the vertex (Figures 4 and 5). During this period, skin cultures were performed, and inflammatory samples (C-reactive protein (CRP), procalcitonin) were analyzed, all with negative results. The mother actively participated in the care process and received instructions on maintaining sterile wound care at home. This included using sterile gloves, compresses, disinfectant, and appropriately sized hydrocolloid dressings. Additionally, the therapeutic approach continued at home through medically assisted video-conferencing.

At 6 months of age, a follow-up cerebral MRI indicated that the bone lesion remained stable in size.

DISCUSSIONS

In most instances (70%), ACC manifests as solitary defects on the scalp. However, there are occasional presentations with multiple lesions. These lesions are typically non-inflammatory and clearly delineated. Despite this, there is considerable debate surrounding the treatment of ACC, primarily due to the remarkably high mortality rates linked to this condition, which range from 20% to 55%.

The heightened rates of mortality and morbidity are attributed to various factors, including:
- Bleeding from the sagittal sinus
- Secondary local infections
- Meningitis
- Thrombosis of the sagittal sinus
- Other significant congenital defects associated with ACC.

Among these complications, bleeding of the superior sagittal sinus poses the most substantial threat to
life, carrying a mortality rate of 36%. This fatal complication typically arises between 1 and 3 months of age and is more prevalent when lesions encompass a larger segment of the superior sagittal sinus. Inadequate safeguarding of the superior sagittal sinus may lead to exposure and subsequent complications. Prompt and efficient management of ACC is imperative to forestall such life-threatening consequences.

The management of ACC involving the scalp remains a topic of debate. Treatment options include conservative and operative approaches, yet no consensus or established guidelines regarding the optimal strategy exist [11]. The decision-making process between conservative and surgical modalities poses a challenge and necessitates individualized assessment for each case. Notably, specialized conservative treatment techniques have been discussed in the literature. Such interventions encompass the utilization of fibroblasts, autologous cultured keratinocytes, and the application of fibroblast growth factors, all of which notably augment wound healing (Figure 7).
CONCLUSION

We maintain that adopting a conservative treatment approach minimizes potential complications that could have arisen from surgical intervention at the lesion site. Remarkably, we achieved rapid skin regeneration within 35 days (Figure 6 and 7). The swift closure of the skin lesion allowed us to exclude the possibility of infectious complications. Looking forward, emerging technologies are positioned to improve wound healing and closure further. Breakthroughs such as fibroblast growth factors and cultured skin may make surgical intervention obsolete.

Conflict of interest:
None declared.

Financial support:
None declared.

Author’s contributions:
Conceptualization, I.F. and M.S.; methodology, G.I.O.; investigation, M.S. and G.I.O.; writing—original draft preparation, M.S. and C.I.G.; writing—review and editing M.S and TV.S. All authors have read and agreed to the published version of the manuscript.

REFERENCES


