Aplasia Cutis Congenita. Case report and literature review

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ABSTRACT
Aplasia Cutis Congenita (ACC) is a rare condition characterized by congenital absence of epidermis, dermis and, in some cases, like this one, subcutaneous tissue and bone. It usually involves the scalp vertex. The estimated incidence is 3 in 10,000 births resulting in a total number of 500 reported cases till nowadays. The lesion can occur on everybody surface, but scalp is the most affected region (70% of the cases). In about 20% of cases there is bone lesion. ACC can occur as an isolated defect or can be associated with a number of other congenital anomalies such as limb anomalies or embryologic malformations. In patients with large scalp and skull defects, there is increased risk of infection and bleeding along with increased mortality and therefore prompt and effective management is advised. Case presentation: We describe a child with ACC involving almost all skull, where it could be seen the brain only with arachnoid membrane. At presentation there was no Cerebral Spinal Fluid (CSF) leakage. And it was managed surgically. The child was operated on one time. It was covered with non-absorbable matrix and the skin was approached. Conclusion: There are multiple treatments for this condition. But there is no consensus on treatment strategies. Conservative treatment can be tried, especially when there is no bone lesion. The treatment must be individualized for each case.

Keywords: Aplasia Cutis Congenita; Congenital Anomalies; Cranial Reconstruction; Scalp Reconstruction

RESUMO
Aplasia Cutis Congênita (ACC) é uma condição rara caracterizada pela ausência congênita de epiderme, derme e, em alguns casos, como este, tecido subcutâneo e osso. Normalmente compromete o escalpo. Estima-se que a incidência seja de 3 em cada 10.000 nascimentos, o que resulta em cerca de 500 casos descritos até hoje. A lesão pode ocorrer em qualquer região do corpo, porém o escalpo é a região mais comprometida (70% dos casos). Cerca de 20% dos casos apresentam lesão óssea. A ACC pode ocorrer como um defeito isolado ou pode ocorrer em associação com outras anomalias congênitas como malformações de membros ou embriológicas. Nos pacientes com grandes lesões no couro cabeludo e comprometimento do crânio, há aumento no risco de infecções e hemorragias com aumento do risco de mortalidade, de forma que é aconselhável o pronto tratamento cirúrgico. Relato de Caso: Descrevemos uma criança com ACC que envolve praticamente todo o crânio, onde observamos o cérebro e a membrana aracnoide. Na apresentação não se observava fistula líquórica (CSF). Foi tratada cirurgicamente. A criança foi operada em tempo cirúrgico único, sendo realizada cobertura com matriz não-absorvível e a pele foi aproximada. Conclusão: Há múltiplos tratamentos para esta condição. Porém, não existe consenso nas estratégias terapêuticas. Pode ser tentado o tratamento conservador, especialmente quando se observa lesão óssea. O tratamento deve ser individualizado em cada caso.

Palavras-chave: Aplasia cutis congenita; Anomalias congênitas; Reconstrução craniana; Reconstrução de escalo
Aplasia cutis congenita is a heterogeneous group disorder characterized by the absence of the epidermis, dermis, and occasionally subcutaneous tissues or even bone tissue, involving multiple possible body locations. The most common lesion location is the scalp (70%) \(^1\)-\(^3\). It was first described by Cordon in 1767. Frieden, in 1986, described a classification system consisting of 9 main ACC types based on number, location of the lesions and presence or absence of associated deformities\(^4\). It is shown on Table 1.

ACC occurs in 3 cases for 10,000 births. The strongest risk factor is the antithyroid drug Methimazole \(^3\)-\(^9\) which, according to Frieden classification (Box), can be categorized on ACC type 8. However, cases of ACC are extremely limited due to the very low incidence and, therefore, it is not possible to devise completely accurate epidemiological data.

The ACC pathogeny is not well known \(^10\)-\(^14\). Otherwise, it’s already known that multiple factors probably contribute to the development of ACC. According to literature:

- Chromosomal abnormalities\(^15\)-\(^16\), specially BMS\(^17\);
- UBA2 gene and the SUMOylation pathway;
- Trauma\(^18\);
- Amniotic irregularities\(^19\);
- Intrauterine complications, such as vascular or infection complications\(^6\),\(^19\),\(^20\);
- Teratogens such as misoprostol, benzodiazepines, valproic acid, cocaine, methotrexate, ACE inhibitors, methimazole\(^5\),\(^8\),\(^13\),\(^21\)-\(^23\).

The main hypothesis about ACC is that the mechanism behind it lies in tension-induced disruption of the overlying skin occurring at 10-15 weeks of pregnancy. At this time, the brain grow occurs fast along with hair direction and patterning\(^16\). Another model is that premature amniotic membrane rupture and amniotic band formation might be the cause of ACC\(^19\).

Here we report a case of a female baby, 37 weeks of pregnancy. She was C-section born without any mishaps. The mother didn’t perform any prenatal exams. No abnormal family records were reported. At the first evaluation, it was observed that the baby didn’t have frontal, parietal and temporal skin, subcutaneous tissue and the cranial bones. Then, she was sent for neurosurgical evaluation. She was born with 2270g.
After specialized evaluation, it was performed the first surgery. The skin was approached and a non-absorbable matrix was sutured. For the 3 weeks later the epithelial tissue grew over the matrix. Around 6 weeks after the surgery, all matrix was covered by the skin.

**DISCUSSION**

In the majority of cases, about 70%, the ACC manifests as a solitary defect of the scalp, but occasionally, it may be present with an extensive lesion, just like ours. There are controversies concerning the treatment of ACC and there has a great scientific interest due to the extremely mortality, that varies from 20% to 55%.

The high morbidity and mortality is caused by sagittal sinus bleeding, secondary local infection, meningitis, sagittal sinus thrombosis. In this case, we observe that the lesion is close to the superior sagittal sinus, therefore the imminent risk of infection and the bleeding is apparent. The major risk of this condition is the bleeding of the superior sagittal sinus. This fatal complication uses to occur in the first three months. It’s caused by sagittal sinus exposure and insufficient protection.

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**Figure 1.** Anterior view. Note the absence of the skin, subcutaneous tissue and bone tissue.

**Figure 2.** Lateral view.

**Figure 3.** Two weeks post-op. Anterior view.
The management of scalp ACC is controversial. Treatment may be even conservative or operative and there is no consensus or guidelines on treatment strategy. Each case must be individualized. Surgery is the first option when we have large lesions, especially involving the sagittal sinus. Conservative management consists of regular wound cleaning and application of dressings along with the use of systemic antibiotics. This includes physiological saline solution, continuous saline drips, betadine solution, bacitracin ointment and silver sulfadiazine dressings. Today we have lots of specialized adherent wound dressings that offer increased wound healing rates and fatty gauzes. None of these materials proved to be better than the other. In small lesions, the conservative treatment is the first choice. Recently there are some cases of large lesions treated in conservative route with excellent results. There have been reports of even more specialized conservative treatment techniques such as the use of autologous cultured fibroblast growth factors that accelerate wound healing, a modality which might represent the future of ACC treatment drastically decreasing the number of surgically managed cases.

On the other side, surgical management, includes various procedures. Standard surgical care includes primary wound closure, skin grafting (autologous or allografts), local scalp flaps with or without tissue expansion, free flaps, muscle flaps, full-thickness or split-thickness skin grafts, and cranial vault reconstruction using bone grafts. Other techniques such as bipedicle opposing local flaps, rotational flaps or L-shaped flaps, have been presenting good results. But large scalp flaps with single wound repair seems adequate and effective for the majority of ACC lesions. Closure of the bone is necessary, it is achieved through the use of bone grafts.

According to the literature the bone can can be self-regenerated with an impressive speed. Some authors also advocate a delayed cranioplasty operation with good results.

Concluding, the exact moment for surgery must be tailored according to each individual patient and lesion, as well as surgeon's personal preference and expertise. The main complications of surgical treatment include intraoperative hemorrhage and postoperative infections. But postoperative complications should not be a discouraging factor for surgery.

REFERENCES


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Conflicts of interests: nothing to declare.