Review Article

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Acutis Dermal Dysplasia: a review

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Aplasia Dermal Dysplasia (ADD) is an etiologically heterogeneous congenital abnormality having a circumscribed area of absent skin that involves any region of the body mainly in cranial vertex. The condition may involve only the superficial skin but also the subcutaneous tissue, bone, or even the dura. ADD can be isolated or associated with other malformations in the context of a genetic disease. Numerous alterations of skin detectable at birth might be confused with aplasia cutis congenita. Birth trauma, injuries from obstetrical procedures and intrauterine trauma such as from mechanical deliveries are some examples. Histologic analysis can make the proper diagnosis. Here we describe the surgical strategies to treatment of ADD, including patient selection, preoperative preparation, the operative procedure and Post-operative management. A multidisciplinary team including neurosurgery and plastic surgery, among others, is optimal for the treatment of these patients. Nevertheless, intensive care must be undertaken during treatment to detect potential life-threatening complications.

Keyworks: Acutis Dermal Dysplasia, skin, Adams-Oliver syndrome, cranioplasty, plastic surgery

INTRODUCTION

Aplasia Dermal Dysplasia (ADD) is an etiologically heterogeneous congenital abnormality having a circumscribed area of absent skin that involves any region of the body, but the great majority of cases present with defects of the cranial vertex in approximately 80–90% of cases [1-3]. Of these scalp lesions, 70% to 75% are single, 20% are double, and 8% are triple. In the literature, more than 500 cases of varying severity have been reported [4].

The condition may involve only the superficial skin but also the subcutaneous tissue, bone, or even the dura. Notably, dilated scalp veins have been described in many cases of ADD. The shapes of scalp lesions vary considerably. Lesions may be circular with a punched-out appearance, roundish to oval, linear, irregularly delimited, linear, or rhomboid. Their surface characteristics can also vary, and may be a wound area denuded of skin, membranous, scarred, ulcerated and granular with suppuration. The margins and surroundings of the scalp defects are not the same in all instances, with some features that include: sharp, smooth margins; raised, wall-like edges; sloping or step-like borders; rolled margins; there may be surrounding healing tissue, circumferential inflammation, pink-white hairless areas, densely haired edges, and they might also be in the early stages of epithelization, channel-like, angiomatous or displaying a collateral course of veins [5] (Figure 1). ADD can be isolated or associated with other malformations in the context of a genetic disease. Adams-Oliver syndrome (AOS) was described in 1945 [6] as a condition involving ADD, most commonly of the scalp and skull, and terminal transverse limb defects. Congenital heart disease has been reported in 13.4% of cases of AOS [7].

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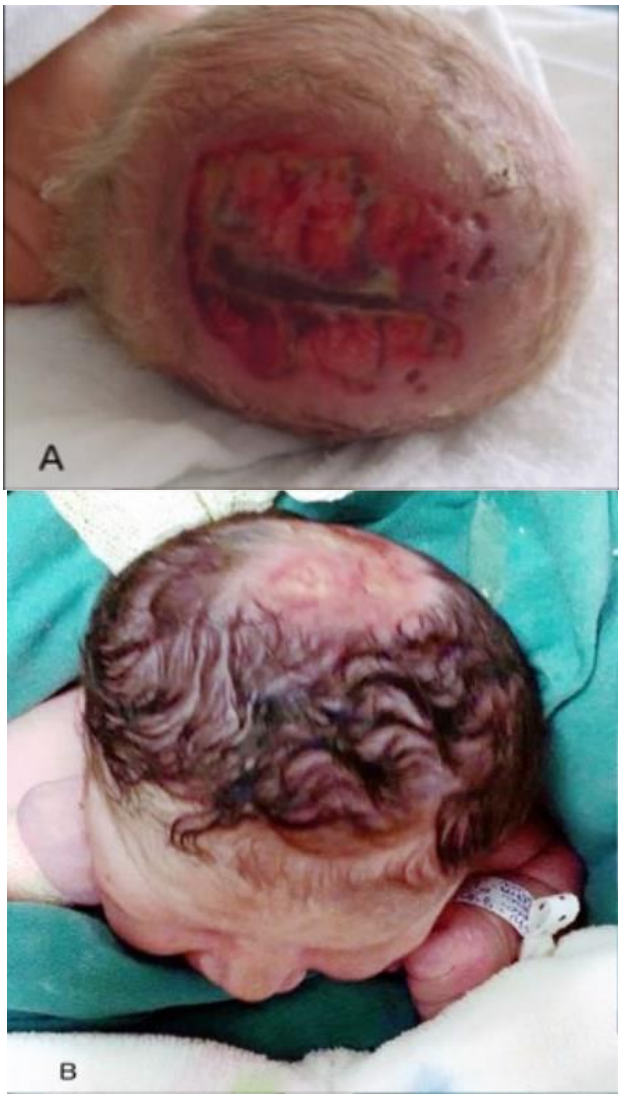

Figure 1 - A Aplasia cutis congenital of the scalp, skull, and dura: the congested sagittal sinus of clearly visible overlining the defect; B. A large scalp and skull defect in the vertex.

Numerous alterations of skin detectable at birth might be confused with aplasia cutis congenita. Birth trauma, injuries from obstetrical procedures and intrauterine trauma such as from mechanical deliveries are some examples. Histologic analysis can make the proper diagnosis [5]. Furthermore, pathologic evaluation of bullous or membranous ADD reveals fibrovascular and/or edematous stroma, similar to the histopathologic appearance of encephaloceles or meningoceles. Histopathology of nonbullous ADD of the scalp shows a layer of thin dermal collagen without overlying epithelium or adnexal structures [5, 8].

PATIENT SELECTION

Although ADD may be suspected antenatally, by means of elevated maternal serum fetoprotein, elevated amniotic fluid fetoprotein, positive amniotic fluid acetylcholinesterase, it is usually diagnosed at cranial inspection soon after birth [9].

The optimal strategy for management of ADD and timing of surgery have been a matter of controversy in the literature. The goal of treatment is to achieve complete closure of the defect avoiding significant risks such as meningitis, hemorrhage and brain injury. Reported treatments include surgical, conservative, or a combination of both [4, 10-13].

Most patients without other associated congenital anomalies have a good overall survival. Limited superficial lesions less than 2 cm wide without exposure of vital structures heal spontaneously when the defect is covered with proper dressings. Defects larger than 2 cm can be treated conservatively or by primary closure, with excellent results [11-13].

In patients with larger scalp and skull defects, treatment may be challenging. The depth of the tissue defect is an important issue. Cases with uncovered cortex are at a higher risk of developing meningitis, sinus thrombosis, and hemorrhage [13, 14]. They may require more aggressive management and early surgical repair. Lastly, general clinical conditions and life expectancy of these babies are also important factors that must be appraised prior to the ultimate therapeutic decision.

PREOPERATIVE PREPARATION

After initial neonatal care, the first step in the management of a patient with confirmed ADD is to cover the lesion with moist dressings to prevent infection and to avoid desiccation and cracking of exposed tissue overlying the dural venous sinus [4, 13]. Genetic studies should be performed as required. Analysis of skull defects is of utmost importance for determining optimal management; therefore, if necessary, plain x-ray, Magnetic Resonance or computed tomography scans should be obtained. Identification of associated congenital anomalies can be achieved by a thorough physical examination; brain scans are used in cases of possible cerebral malformations, which can be present even when the skull is intact.

Conservative management has been advocated by several authors [4] (Figure 2). The rationale for this approach is to avoid the risks of surgery. Patients are treated with daily silver sulfadiazine or bacitracin ointment dressings in order to reduce infection rates and dehydratation of the wound (iodine dressings should be avoided because they can lead to drying of the scar and bleeding from the sagittal sinus). If successful, conservative treatment could be maintained until healing is complete. Nonetheless, there are some reported

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Figure 2 - (A) Patient with a large defect in the scalp and cranial vault at the vertex at the age of 2 days. (B) Four months later after conservative management. (C) Complete closure of the wound at 6 months of age. (D) A CT scan showed a huge lack of bone at 2 years of age

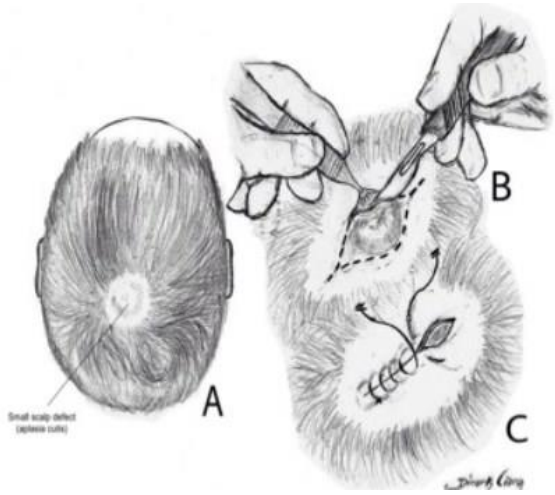

Figure 3 - (A) Artist's sketch of scalp showing a small cutis dermal dysplasia defect. (B,C) Primary skin closure.

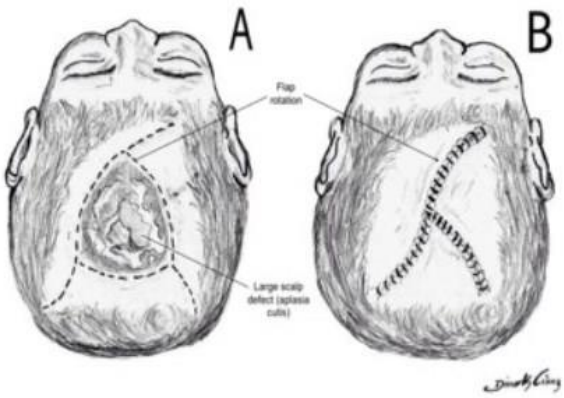

Figure 4 - (A) Artist's sketch of scalp showing a cutis dermal dysplasia involving the midline with a sagital sinus exposure. (B) the scarred area was excised. Margins were approximated after wide undercutting with the aid of multiple sub-cutaneous relaxation incisions as indicated by the dotted lines. A flap rotation was used to cover the skin defect.

cases of ADD with fatal evolution after massive hemorrhage in patients conservatively treated [13, 14].

OPERATIVE PROCEDURE

Given its widely variable clinical presentation, there is no standard surgical strategy for ADD and surgery must have tailored on an individual basis. Small lesions with normal pericranium and skull can be closed primarily, after the subgaleal plane has been widely undermined. Surgical treatment options for larger defects include split or full thickness skin grafts, scalp rotation flaps, pericranial flaps, split rib grafts with a latissimus dorsi muscle flap and tissue expansion [13]. In such cases, plastic surgery consultation is advised. Tissue engineering technology has been brought to bear on this condition as well, with successful descriptions of skin coverage by successive grafts of cultured fibroblasts and/or epidermal cells [15] (Figures 3 and 4).

Nevertheless, there is a high risk of partial or total flap failure when large flaps are used because of their size and associated abnormalities of the adjacent skin, leading to insufficient vascular supply. Flap reconstruction also involves prolonged general anesthesia and a major surgical procedure, with the risk of significant blood loss and complications. Secondary hemorrhage (from the superior

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sagittal sinus) and infection are the two most severe pitfalls in ADD. Mortality rates range from 25% to 55% [4, 13].

POST-OPERATIVE MANAGEMENT – CRANIAL DEFECTS

The sizes of cranial defects vary widely, and, except in rare reports, the bony defects are usually smaller than the overlying scalp defects. Bone grafting at the time of closure of the neonatal scalp should rarely if ever be done, because complete or nearly complete ossification commonly follows early closure of the scalp. Complete bony closure is also reported with nonsurgical management [4]. Persistent bony defects of critical size may require cranial reconstruction, but this is inadvisable before about 3–4 years of age because (a) splitting the very thin neonatal cranial bone can be difficult or impossible due to the thinness of the bone, which has little or no cancellous diploe, and (b) the risks probably outweigh possible benefits [13].

CONCLUSIONS

Conservative management in cases of ADD is safe and that good outcomes can be expected as compared to those managed surgically, also avoiding the risks of complications related to the operative procedure and to unfeasible skin flaps or heterologous grafts. These cases do not necessarily imply that the complications of hemorrhage, sagittal sinus thrombosis, meningitis, brain hernia, and hydrocephalus can be prevented by this method. Flap rotation and other techniques have a major role in this context, but they should be used only after careful consideration or as a salvage procedure.

A multidisciplinary team including neurosurgery and plastic surgery, among others, is optimal for the treatment of these patients. Obviously, either for conservative of surgical treatment, parents should be informed of the potential complications involved. Late surgery and calvarial bone reconstruction might be recommended later for cosmetic reasons. Nevertheless, intensive care must be undertaken during treatment to detect potential life-threatening complications.

DISCLOSURES

Ethical approval

This study was performed in line with the principles of the Declaration of Helsinki. Approval was granted by the local Ethics Committee

Consent to participate

The patient gave consent to use his information and images for research proposes.

Consent for publication

The patient gave consent to use his information and images for publication.

Conflict of interest

The authors declare no conflicts of interest with respect to the content, authorship, and/or publication of this article.

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