Management of large scalp and skull defects in a severe case of Adams-Oliver syndrome

Case report

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Adams-Oliver syndrome is a rare congenital disorder that includes congenital scalp and skull defects, variable degrees of terminal transverse limb anomalies, and cardiac malformations.14 Cutis aplasia occurring in 75% of patients is a potentially life-threatening condition. Large skin defects that cannot be closed primarily present a management dilemma, and may require skin grafting or flaps, or a combination of both operative and conservative modalities. The authors’ experience in management of huge scalp and bone defects with the Integra Dermal Regeneration Template and regular dressing changes showed good scalp repair and no serious complications attributed to this approach. (DOI: 10.3171/2009.7.PEDS09220)

Key Words • Adams-Oliver syndrome • scalp defect • skull defect • synthetic dermal graft

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dams-Oliver syndrome is a rare congenital disorder first described in 1945, which includes congenital scalp and skull defects, variable degrees of terminal transverse limb anomalies, and cardiac malformations.14 Cutis aplasia occurs in 75% of patients with AOS that involves the cranial vertex, and can be extensive, exposing the dura mater and deeper meninges.15 There is no established cause for cutis aplasia. Although amniotic adhesions, intrauterine pressure, and hereditary factors have been suggested, none of these (except heredity), has been confirmed to be the cause.7,18

We report a case of AOS in which cutis aplasia involved a large portion of the skull. The day after birth, the patient was treated with early synthetic dermal substitute replacement due to CSF leakage and to prevent infection. Her defect was managed conservatively with repeated dressings. This case report is presented to highlight the steps of one method of successfully managing large cutis aplasia.

Abbreviation used in this paper: AOS = Adams-Oliver syndrome.

Case Report

History and Examination. This 1-day-old girl, the product of full-term pregnancy from a healthy 35-year-old mother, was referred with large congenital scalp and skull defects. The pregnancy had been normal except for a prenatal diagnosis of occipital encephalocele, for which the mother declined amniocentesis for further investigation. The child was born by cesarean section after spontaneous rupture of the membranes at another medical center. Apgar scores were 8 and 8 at 1 and 5 minutes, respectively. The neonate was transferred shortly after initial medical stabilization, with normal saline-soaked sterile gauze placed on the occipital defect. She had a 10 × 12–cm rectangular midline occipital defect with visible brain tissue, demonstrating a complete lack of scalp soft tissue and cranial bone, which was associated with
ruptured dura mater and leakage of CSF (Fig. 1). Other obvious abnormalities included an anterior midline scalp skin tag at the region of the anterior fontanel; a midline midthoracic spine skin tag; and anomalies of both lower extremities, with left clubfoot, 3 toes on the left foot and 4 toes on the right, fingertip amputation, and syndactyly of the hands (Fig. 2).

Operation and Postoperative Course. Cranial ultrasonography studies obtained immediately after birth revealed a cystic structure in the posterior fossa and ventriculomegaly. After medical stabilization of the neonate, an operation was performed by the neurosurgery and plastic surgery teams on the 2nd day of life. The dural defect was closed primarily with braided nylon (Neurolon) suture. The skin defect was covered with Integra Dermal Regeneration Template (Integra Life Sciences) (Fig. 3, Table 1). Postoperatively the infant was taken to the neonatal intensive care unit and underwent sterile dressing changes and an intravenous antibiotics regimen for 10 days. Magnetic resonance imaging and CT scanning of the brain revealed corpus callosum agenesis as well as cortical dysplasia and intraventricular hemorrhage with severe hydrocephalus (Fig. 4). On postoperative Day 14, a right frontal access device was inserted to manage the intraventricular hemorrhage and CSF leakage from the skin edges in the occipital area. Formal ventriculoperitoneal shunt insertion was performed 2 weeks later, subsequent to resolution of the hemorrhage.

There was no history of similar abnormal findings in the patient’s relatives. Additional workup related to her symptoms that were compatible with AOS was performed. Results of a chromosomal study were unremarkable. Spinal MR imaging showed dysraphism at the level of T3–4, and cord tethering with a small dorsal tract extending from the subcutaneous tissue and through a bone defect to the spinal canal. The echocardiogram obtained 1 week after birth was significant for atrial septal defect (secundum).

Seven days after birth, granulation tissue at the periphery of the scalp defect started to form, and the defect progressively became smaller as stable granulating soft tissue associated with epithelialization continued to migrate toward the center of the defect. The child was discharged home at 1 month of age, with regular dressing changes for the reduced scalp defect. She was readmitted 2 weeks after

<table>
<thead>
<tr>
<th>TABLE 1: Composition, properties, and contraindication of synthetic dermal template</th>
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<tr>
<td>Integra Dermal Regeneration Template</td>
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<tr>
<td>composed of a porous matrix of cross-linked type I bovine tendon collagen &amp; glycosaminoglycan, &amp; a semipermeable polysiloxane (silicone layer)</td>
</tr>
<tr>
<td>provides immediate wound coverage</td>
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<tr>
<td>provides the needed framework for blood vessels &amp; dermal skin cells to remodel damaged site</td>
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<tr>
<td>as skin cells migrate into the matrix, collagen is slowly absorbed &amp; replaced w/ collagen produced from the person’s own cells; complete wound closure occurs as epidermal cells migrate from wound edges</td>
</tr>
<tr>
<td>should not be used in patients w/ known sensitivity to bovine collagen or chondroitin materials</td>
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discharge with skin maceration overlying the right frontal access device. The CSF culture confirmed bacterial infection, and the ventriculoperitoneal shunt and the frontal device were removed. A new shunt was inserted after the child was treated successfully with broad-spectrum antibiotics.

Only 6 weeks after birth, the child demonstrated a minimal scalp defect after continued conservative dressing management. At 13 months of age, the patient has developed complete soft-tissue coverage in the occipital region (Fig. 5). There is no focal motor deficit. Her pediatric follow-up findings are appropriate, with only mild delay in developmental milestones. Her most recent CT and MR imaging studies reveal control of hydrocephalus and the presence of acceptable scalp and bone coverage of a previous large congenital defect (Fig. 6).

**Discussion**

Adams-Oliver syndrome manifests variably, with a wide range of scalp defects and limb malformations. Scalp deformities frequently involve the vertex, and sometimes are associated with skull defects. Limb defects can differ from nail dystrophy to complete absence of distal extremities, most frequently observed as syndactyly, brachydactyly, polydactyly, oligodactyly, and hypoplastic fingernails and/or toenails. Associated features include a spectrum of anomalies ranging from skin tags to lymphedema, cardiac malformations, and brain abnormalities (microcephaly, epilepsy, mental retardation, Dandy-Walker malformation, and hydrocephalus).1,5,6,20

Both autosomal-dominant and recessive inheritance have been found in afflicted patients. Most reported cases are sporadic; however, familial occurrence has also been described. The sporadic cases may characterize incomplete penetrance of the autosomal-dominant mode of transmission.4,14 Our patient had no known family history of congenital deformities of the scalp and extremities, and results of her chromosomal study were normal.

Although a clear pathophysiological mechanism has not been recognized for AOS, vascular compromise in watershed areas, with or without secondary amniotic bands or external compression during pregnancy, has been suggested as a possible origin for AOS.10,14

A multidisciplinary approach to the symptomatic af-
fected organs in AOS is advised. Survival is compromised because of associated pathological conditions, including hydrocephalus, cardiopathies, and hydronephrosis.3,4

Management of cutis aplasia has been controversial because AOS is a rare condition and there is a lack of extensive clinical experience related to outcomes of differential therapeutic modalities. The goal of treatment is to achieve complete closure of the defect, avoiding major complications such as meningitis, hemorrhages, and trauma to the brain.3,4,18 Reported treatments have included surgical closure, conservative management, or a combination of the two.3,13,17 Determination of appropriate clinical management during the early stages of life improves survival and is based on the size of the defect, presence of an underlying skull defect, the child’s general condition, and his/her associated life expectancy.3,18

Conservative treatment is advocated by some authors because of its simplicity.3,16 As initial management of the condition in newborns, dressing changes avoid the possibility of surgical complications and the potential failure of skin flap or heterologous graft coverage.2 Conservative treatment of cutis aplasia also maintains dural induction of osseous regeneration.16 Typical dressing regimens in conservative treatment include daily wound care with moist sterile gauze and topical application of the antimicrobial agents silver sulfadiazine or bacitracin ointment, in conjunction with a systemic antibiotic therapy for the first several weeks, treatments that reduce the bacterial load and avoid dehydration of the wound.4,7,18 Use of non-adherent, atraumatic, and relatively cheap dressings such as Mepitel has reportedly been successful for the initial conservative treatment to manage large defects in AOS.12

Conservative therapy complications include meningitis, significant hyponatremia with seizures, and brain herniation. Death after massive hemorrhage in patients undergoing conservative treatment also has been reported.4,9,18 Povidone-iodine dressings should be avoided because they lead to wound desiccation, with the potentially fatal complication of bleeding from the sagittal sinus.18 Late surgery and bone reconstruction associated with alopecia and residual calvarial scarring may be indicated at a later point for aesthetic reasons.

For large lesions with an underlying skull defect and uncovered brain, CSF leakage, and uncommon dural blood vessels like an arteriovenous fistula, surgical treatment is recommended to prevent massive hemorrhage and a high mortality rate of ~20% secondary to infection, sinus thrombosis, or hemorrhage from ulceration of the sagittal sinus.2,4,18 However, it remains unclear how to treat patients with a large defect without the aforementioned risk factors. Apart from an elevated surgery-related risk of hemorrhage and perioperative infection in infants, any graft, especially if it covers large defects, involves a high risk of partial or total graft failure because of the size of the defect or the associated abnormality of the adjacent skin.4

There is a lack of consensus about the size of scalp defects appropriate for surgery. Some advise that sizes > 1 cm should be treated surgically.3 Others elect to treat lesions > 2 cm conservatively or by primary closure, with excellent outcome.18,19 Ross et al.17 recommended skin grafts for defects with a diameter of > 5 cm. These authors recommended a combined regimen of conservative and surgical treatment for cutis aplasia to decrease the complications of conservative treatment and the chance of unsuccessful surgical treatment in patients with a large defect.

Surgical regimens for cutis aplasia have included primary closure if possible, split-thickness or full-thickness skin graft, scalp rotation flaps, pericranial flaps,7 split rib grafts with a latissimus dorsi muscle flap in which tissue expansion was used,19 and a 1-stage operation of free tissue transplantation (free flap) performed using microsurgical vascular anastomosis.11,16,18 In the AOS, large rotation scalp flaps are not reliable due to the abnormal vascularity of the skin.16 Challenges associated with operative therapy include poor soft-tissue coverage, donor site morbidity, difficult separation of the underlying brain from the transplanted covering in secondary operations, persistent alopecia, and potential brain damage. In addition to the significant perioperative risks, surgical procedures may inhibit the osteogenic potential of the dura mater, which usually results in poor regrowth of bone.3,16 There is an equally high mortality rate (20%) associated with conservative treatment and with surgical repair.4

The scalp and skull defect in our patient was managed immediately (the day after birth) with dural repair and coverage of the large defect with synthetic dermal substitute, which provided adequate coverage. Potentially life-threatening complications in this instance were avoided with prompt CSF drainage and appropriate shunt management. Repeated dressing changes were the mainstay following graft placement to foster good wound healing and to limit the need for further surgical intervention.

Conclusions

Cutis aplasia in AOS is a potentially life-threatening condition. In general, there is agreement on the proper management of small lesions in which the brain is covered. Large skin defects that cannot be closed primarily present a management dilemma, however, and may require skin grafting or flaps, or a combination of both operative and conservative modalities. Our experience with synthetic graft and regular dressing changes showed good scalp repair and no serious complications.

Disclaimer

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

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