Rare disease

Two different management modalities in a two sibling case report of Adams Oliver syndrome

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Correspondence to Miss Nada Al-Hadithy, nadaucl@yahoo.com**Summary**

The authors report two-sibling case of Adams Oliver syndrome. One was managed with early surgical intervention after a period of failed conservative management and the other conservatively managed until reconstruction and surgical correction of alopecia. The goal of treatment is to achieve a complete closure of the defect without encountering any fatal complications such as haemorrhage and infection or causing any long-term neurological morbidity. Various papers have been published related to ACC, yet there is no consensus on the therapeutic approach. The management decision hinges upon balancing the risks of complications including spontaneous sagittal sinus haemorrhage and the risk of surgical intervention. Both treatment modalities proved successful in these cases.

BACKGROUND

Aplasia cutis congenita (ACC) is a rare condition, part of a heterogeneous group of disorders first reported by Cordon in 1767 and is characterised by the absence of a portion of skin in a localised or widespread area at birth. It most commonly (70%) manifests as a solitary defect on the scalp, but sometimes it may occur as multiple lesions and can be associated with other physical anomalies or malformation syndromes. In 75% of the cases, the defects are small (less than 2 cm) and confined to the skin.¹ Larger lesions may be associated with aplasia of the underlying skull, and in these cases the mortality has been estimated up to 60%.² Patients with such defects are at significant risk for sagittal sinus haemorrhage, venous thrombosis, desiccation and infection of the meninges, problems with thermoregulation and electrolyte abnormalities with the risk of seizure.²⁻³

Frieden created a classification system for ACC consisting of nine groups based on the number and location of the lesions and the presence or absence of associated malformations.⁴ In 1945 Adams and Oliver first reported the association of cutis aplasia with terminal limb anomalies.⁵ Adams Oliver syndrome (AOS) is Frieden's Classification Group 2.⁶

We present two cases of phenotypically similar AOS, with large full thickness cutis aplasia associated with cardiac and limb anomalies.

CASE PRESENTATION

Case 1 A 1-day-old 41 weeks girl weighing 3250 g with a large area of cutis aplasia was presented to our unit following spontaneous vaginal delivery. There was a 15×9 cm full thickness defect of the scalp and calvaria with exposed dura overlying the sagittal sinus. Apgar scores: 2 at 1 min and 7 at 5 min and required assisted ventilation with a bag-valve-mask device for 1 min. She suffered left focal seizures at 12 h and 15 h of age each lasting 1 min. Further clinical examination demonstrated a bicuspid aortic valve and mild pulmonary stenosis, absent toenails of the left foot, absent distal phalanges of the left foot and brachydactyly of both

feet and absent ossification of middle and distal phalanges of the left foot. This was followed up by cardiologists and orthopaedic surgeons.

Family history included a 1 cm scalp defect and bicuspid aortic valve with aortic stenosis in her father. He had no anomalies involving his extremities. A paternal uncle had been born with a hypoplastic left heart and died at the age of 5 days, and another sibling with coarctation of the aorta. The infant's mother had no significant family history relating to cutis aplasia, cardiac, or extremity malformations.

Figure 1 Clinical photograph of case 1 with tissue expanders inserted.

She was managed conservatively with mepitel dressings and antibiotics: benzylpenicillin, gentamicin and metronidazole and a loading dose of phenobarbiton. She was discharged 12 days later with normal neurology. She was managed conservatively with dressings. At age five, there was still a significant calvarial defect and she underwent definitive treatment with craniotomy, split calvarial grafts and medial advancement of expanded skin (figure 1). She has made an uneventful recovery reaching normal milestones with neurological defect observed.

Case 2 A boy was born at 40+ 6 weeks, weighing 3210 g following an induced labour, in an otherwise normal vaginal delivery. No maternal medication was taken during the pregnancy and there was no evidence of infections with varicella or herpes viruses. Antenatal screening had been carried out with ultrasound which demonstrated a scalp defect.⁴

Figure 2 Clinical photograph of case 2 with a haemorrhagic lesions on the vertex of his scalp.

An extensive, 12×9 cm midline defect was noted at birth. This was a full thickness cutis aplasia over the posterior parietal region, resulting in an exposed sagittal sinus and only meningeal covering over brain. A small 1 cm haematoma was noted at the 1'o clock position. Neurological examination was unremarkable. He was also noted to have cutis marmorata telangiectatica and a 2–3 toe syndactyl on left foot – with atrophic small toes. There were no cardiac anomalies.

He was transferred to our care on day 1 for conservative management with mepitel dressings and intravenous ceftriazone. By day 8 the initially small vascular lesion on the perimeter had enlarged into a large haematoma and

Figure 3 MRI T1 sagittal through the brain demonstrating encephalocele.

Figure 4 Management options in partial thickness cutis aplasia.

Figure 5 Management option in full thickness cutis aplasia.

was actively bleeding (figure 2). Preoperative MRI revealed that the haematoma was in fact a necrotic encephalocele (figure 3). This was surgically debrided on the same day. A duragen patch was used to cover the residual dural surface defect and a fascial lata graft was taken from the left thigh. His head was dressed with mepitel, wet swabs and a dry hat. He was kept ventilated overnight to minimise intracranial pressure and followed up with a full septic screen including lumbar puncture. He was discharged with grossly normal neurology on day 25. On follow-up the fascial lata graft had taken 100% and he has continued to reach developmental milestones appropriately.

Depending on the extent of osseous regenerative, definitive cranial reconstruction with split calvarial grafts and tissue expansion are planned for when he was old enough to comply with treatment.

INVESTIGATIONS

- ▶ Septic screen with bloods and microbiology swabs
- ▶ MRI
- ▶ Lumbar puncture
- ▶ Echocardiogram
- ▶ Skeletal survey with x-rays.

OUTCOME AND FOLLOW-UP

In both cases there were no neurological complications or developmental delay.

DISCUSSION

AOS is a distinct subtype of Frieden's Classification Group 2.⁷ Most AOS cases are sporadic but there is a definite genetic element that may run in families, in these cases there is an autosomal dominant pattern of inheritance with a variable phenotypic expression.⁸ The scalp lesions tend to be large. The most common limb malformation

is hypoplastic or absent distal phalanges. Other anomalies may include cutis marmorata telangiectatica congenita, haemangiomas, cranial arteriovenous malformation, skin tags, supernumerary nipples, woolly hair, congenital cardiac anomalies. Congenital heart defects can also be present in about 12% of cases.

The mechanism is thought to be due to a disruption of the blood flow in small vessels as an actual abnormality of small vessel development in the early embryo rather than these vessels being clogged by clots.^{9 10}

The goal of treatment is to achieve a complete closure of the defect without encountering any fatal complications such as haemorrhage from the underlying dural venous sinuses, local and central nervous system infection or causing any long-term neurological morbidity. In the long-term, reconstitution of the bony calvaria and scalp with protection of the underlying brain, and appropriate cosmesis and hair growth is desirable. Various papers have been published related to ACC, yet there is no consensus on the therapeutic approach.¹¹ The management decision hinges upon balancing the risks of spontaneous haemorrhage from the risk of surgical intervention in different sizes of lesions and varying degrees of depth of defect. Small lesions less than 2 cm are usually not associated with calvarial defects and can be managed conservatively (figure 4).

In larger lesions, exposed dura can scar and dry out. Subsequent separation of scar tissue from the wound bed can cause spontaneous haemorrhage. Therefore, early surgical intervention is indicated in ACC with involvement of underlying dura and the brain, early eschar formation, massive haemorrhage, cerebrospinal fluid leakage and arteriovenous fistula.^{12 13} If there is a large (>5 cm) calvarial defect in a child older than 12 months, enough ossification to fill the defect is unlikely.¹⁴ Thus, surgical treatment can be used to replace and to heal bony defects in addition to cutaneous

defects, either as an immediate or delayed intervention. Surgical treatment options include skin grafts, local flaps with or without tissue expansion and free flaps. A summary of treatment algorithms can be seen below (figure 5).

Cranioplasty may be performed immediately or late.¹⁵ If repaired, calvarial defects can be reconstructed autologously or with alloplastic material.

Surgical debridement of the scar tissue may also induce a fatal bleeding from the sagittal sinus. Additionally the risks of surgery include poor soft tissue coverage, flap necrosis, graft loss, donor site morbidity, potential brain damage and porencephalic cyst. Skin grafts may interfere with the expansion of neurocranium. Additionally surgical procedures may inhibit the osteogenic potential of the dura to initiate and sustain bony closure of the defect.¹²

Due to these risk factors conservative treatment has been advocated by several authors.¹⁶ An optimal conservative management plan has to provide protection against infection, keep the wound sufficiently moist and supply the optimum wound environment.¹⁷ An example of this would be Omiderm (Taureon, The Netherlands),¹³ which is made of a permeable polyurethane film which minimises fluid loss, but is permeable to oxygen and antibiotic solutions and prevents microbial invasion. In the literature, non-adherent topical antibacterial dressings such as silver containing Acticoat (Smith & Nephew, St Petersburg, Florida, USA)¹⁸ and Elasto-Gel (Southwest Technologies Inc, Kansas City, Missouri, USA) offer a shorter healing time, and reduced need for dressing changes, thereby protects from shock and friction.¹⁹

Povidon iodine and bacitracin have been suggested with or without systemic antibiotics for protection against infection.²⁰ However can cause drying and separation of scar tissue with the associated risk of inducing fatal haemorrhage of the sagittal sinus.²¹ Other authors have advocated against the use of systemic or topic antibiotics because of the risk of cytotoxicity, high risk of resistance and the incidence of allergic contact dermatitis.²²

Both of our cases were larger than 2 cm and therefore, according to many authors should have been treated with surgery in the first instance. However, we believe that these cases demonstrate conservative management that can work in larger defects. We recommended an initial attempt with conservative management using new wound dressing materials to avoid possible complications from surgical procedures as well as reducing donor site morbidity. The patient and parents must be counseled regarding the complications such as massive haemorrhage, cerebrospinal fluid leakage or an arteriovenous fistula, in which surgical intervention may be necessary.

SUMMARY

We present the surgical management of two patients with AOS and large full thickness osseocutaneous defects of the vertex of the scalp. Surgical management included delayed split calvarial grafts and tissue expansion and advancement, and primary fascial lata graft in the case of a haemorrhagic encephalocoele. These treatments resulted in no long-term neurological morbidity and satisfactory cosmesis with coverage of alopecia.

Learning points

- Patients presenting with cutis aplasia must be examined from top to tail to identify any associated limb or cardiac anomalies.
- In large defects, the mortality from cutis aplasia can be up to 50%.
- Management of cutis aplasia can be conservative or surgical and must be pragmatically tailored to the clinical indications.

Competing interests None.

Patient consent Obtained.

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