Adams-Oliver syndrome, a successful conservative approach for a large scalp defect

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Abstract

Adams-Oliver syndrome was first described in 1945 as a multiple congenital malformations association including aplasia cutis congenita and terminal transverse limb defects, along with cardiovascular and central nervous system anomalies. We report the case of a boy, prenatally diagnosed with a malformation of feet and right hand. At birth, a malformation of the skull was observed, at midline and right frontal, parietal and occipital region, with meningeal exposition. He presented with abnormal feet and right hand with hypoplastic fingers and also exhibiting cutis marmorata telangiectatica. Cardiac, abdominal and central nervous system malformations were excluded.

He started a conservative approach based on daily dressings. The scalp defect closed at 4 months with this management strategy. At this age, a skull defect about 5 cm long was still perceptible by palpation of the area. The boy showed normal growth and neurologic development. No complications were reported.

This report reinforces the effectiveness of conservative management strategies for extensive bone and epithelization defects in syndromes of aplasia cutis congenita like Adams-Oliver syndrome.

Keywords

Adams-Oliver syndrome, aplasia cutis congenita, conservative treatment, topical therapy, congenital scalp defect, limb defects.

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How to cite

Introduction

Adams-Oliver syndrome (AOS [MIM100300]) was first described in 1945, as a multiple congenital malformations association, including aplasia cutis congenita and terminal transverse limb defects, often associated with cardiovascular and abdominal wall defects [1, 2]. Central nervous system anomalies, with variable involvement of brain and eyes, and others, are also reported [3, 4].

A wide phenotypic spectrum is possible. Descriptions diverge from minor cutis defects as an asymmetric skull shape with facial peculiarities along with syndactyly, to a gross cerebral malformation leading to developmental delay with epilepsy or visual impairment. An individualized and comprehensive multidisciplinary approach is always necessary.

In this report we intend to describe the malformations of a boy with Adams-Oliver syndrome, with a large scalp and skull defect, and also the conservative management strategy used, with success.

Case report

We report the case of a boy, the second son of young, healthy and non-consanguineous parents, with a healthy first child. His maternal great-grandfather was born with a small defect in occipital scalp that was only perceptible when adult as a tiny alopecia area.

The boy had a diagnosis of malformation of both feet and right hand since the 25th gestational week, apparently lacking fingers (Fig. 1 and Fig. 2). The fetal karyotype was normal (46XY).

The newborn was delivered at 39 weeks of gestational age, by vaginal route, with Apgar score 10 at 1st and 10th minutes. The birth weight was 3.475 kg (50th-90th percentile), length 51 cm (50th percentile) and head circumference 34.5 cm (10th-50th percentiles).

Immediately after birth, a huge defect involving scalp and skull was observed, at midline and right frontal, parietal and occipital region, about 12 x 5 cm long. It was possible to observe the brain through a thin, translucent and intact membrane, that rapidly became dark (Fig. 3). He also presented abnormal feet and right hand, with hypoplastic fingers, along with cutis marmorata telangiectatica (Fig. 4). The X-ray showed a right hand with hypoplastic bones and agenesis of distal phalanges. The feet showed similar aberrations with atrophic phalanges and small metatarsal bones (Fig. 5).

Remaining physical – including neurological – examination were normal.

The workup study performed at birth excluded cardiac, abdominal and central nervous system malformations. Brain CT showed a long defect of frontal parietal and occipital skull, at midline and right side, about 12 x 6.5 cm long (Fig. 6).

The boy was admitted in our Neonatal Intensive Care Unit (NICU) for intravenous antibiotics during seven days. He maintained a good general condition, with vigorous suction and normal archaic reflexes. Infectious biochemical markers were negative.

During his stay, he started a conservative restorative treatment of the scalp defect that included...
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silver covered by hydrogel. This strategy had the advantage of allowing hospital visits to happen each 3 days. The epithelization continued and at the 4th month of treatment there was only a small and linear skin defect that solved in a week with a final dressing including maltodextrin. Topic betamethasone was used along all the process, intermittently for cycles of 2 or 3 days, when hypergranulation areas arise.

At 4 months, the scalp defect was closed but a skull defect about 5 cm long was still perceptible palpating the area (Fig. 7).

The boy is currently 12 months. The skull defect is still noticeable (about 3 per 4 cm).

He doesn’t walk yet but is capable of standing with some support and using appropriate shoe inserts. He shows normal growth and neurologic development,
without any complications. He maintains a multidisciplinary follow-up plan, including physical rehabilitation, as an attempt to anticipate and overcome the probable motor difficulties that will come up. A genetic appointment is also scheduled.

Discussion

Since Adams and Oliver first described 8 members of a family with aplasia cutis congenita and terminal limb defects, several reports of this syndrome with a large spectrum of congenital anomalies have been described [1].

The etiology of AOS is not completely clarified. Specific genetic mutations have been recently reported in literature, associated to autosomal dominant or recessive forms [5]. Most are transmitted as a dominant trait, like the ones concerning genes ARHGAP31, RBPJ or NOTCH1 [6-8]. Some show recessive transmission, like the ones affecting genes DOCK6 or EOGT [3, 9]. It is possible that other genes not yet identified could also be responsible for the syndrome.

In our report, parental examination was normal but there is family history that a maternal great-grandfather was born with a diminutive defect in occipital scalp, still perceptible when adult as a small alopecia area. That points towards a familial occurrence. The patient was referred to genetic consultation and a familial genetic study was hypothesized.

The malformation of feet and right hand are the most common in this syndrome, with an estimated prevalence of 84% [4]. As reported, our patient had abnormal feet and right hand, with hypoplastic fingers and atrophic nails. So far, they have had no specific treatment but a close follow-up including functional adaptations will be important through his growth.

Aplasia cutis congenita is the second most frequent defect, present in about 75% [4]. Our report shows a large defect in scalp and skull, with successful reepithelialization on a conservative management. There are lots of controversies regarding the best strategy to manage the skull defects resulting from aplasia cutis congenita. The uncovered brain is at great risk of infection, hemorrhage and thrombosis of the superior sinus, or herniation, as reported by Tröbs et al. [10].

Surgical treatment ranges from simple closure to scalp flaps or tissue expansion when possible. A split rib cranioplasty was used to reconstruct the cranial vault in aplasia cutis congenita. However, probably because of the abnormal skin with an atypical vascularization, these procedures have high rates of failure [11].

Conservative treatment, due to the ability of rapid regeneration in the newborn and the osteogenic potential of the intact leptomeningeal covering, has been described as the preferred initial treatment [10-12]. The goal of conservative treatment is to allow healing by secondary intention whilst avoiding infection, dessication and eschar formation. However, this process is time consuming, and complete osseous regeneration may take 1 year [12]. Şen et al. reported a successful conservative management with silver sulfadiazine and gauze dressing after failed surgical treatment [11]. Santos De Oliveira et al. recommended daily dressings with silver sulfadiazine or bacitracin cream [12].

We first used silver sulfadiazine cream; at the third month it was exchanged with nanocrystalline silver, which has also antimicrobial properties and healing promoter capacity and allowed the dressing to be changed only every third day instead of daily. Finally, when the defect was minor, we used a dressing made of maltodextrin, which has antibacterial and wound healing properties, based on our experience with this product in the treatment of pressure ulcers.

With this management strategy, the child was hospitalized only for 7 days at birth. He needed daily visiting to the hospital until 3 months and after this every 3 days. At 4th month, the scalp defect had completely epithelialized, with no complications.
The cranial vault defect has also evolved favorably, and the skull defect at 4 months was more than 50% smaller than at birth. At first year only a small defect similar to a large fontanelle is perceptible.

Another clinical feature of Adams-Oliver syndrome is cutis marmorata telangiectatica, in about 12% to 25% of cases [4]. It’s a venous or capillary malformation that in our patient was generalized.

Previous reports mention many other malformations, like cardiac in 13% to 20%, or from central nervous system, leading to developmental delay or seizures. Renal, genito-urinary, ophthalmological, hematological and pulmonary anomalies are described as well [4]. To date no other anomalies besides the reported were found in our patient.

Because of all this particularities, a close comprehensive multidisciplinary approach will be necessary.

Conclusions

This case illustrates a successful conservative approach for a large defect of scalp and skull in Adams-Oliver syndrome. The best management strategy is still a controversy, but surgical treatments, much more aggressive and expensive, have not shown better results and have high rates of failure.

So far there have been no complications, namely infection, hemorrhage, thrombosis or herniation, or others like convulsions.

We believe that the prognosis in our case will be favourable since no other major malformations like cerebral alterations were found, but a tight follow-up will be necessary.

Declaration of interest

The Authors have no conflicts of interest to declare.

References