

## Adams-Oliver syndrome: a case with full expression

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### Abstract

Adams-Oliver syndrome (AOS) is characterized by the combination of congenital scalp defects (aplasia cutis congenita) and terminal transverse limb defects of variable severity. It is believed that Adams-Oliver syndrome without major organ abnormalities does not necessarily alter the normal lifespan. We present a case without detectable major organ abnormality contrary to life but with poor weight gain. A male infant with scalp and skin cutis aplasia, generalized cutis aplasia, dilated veins over scalp and trunk, hypoplastic toes and nails of feet, glaucoma, poor feeding and poor weight gain. This report shows a case of AOS without major multiple organ abnormalities but with poor feeding and abnormal weight gain that may be alter the normal lifespan.

### Introduction

Adams-Oliver syndrome (AOS) is characterized by the combination of congenital scalp defects (aplasia cutis congenita) and terminal transverse limb defects of variable severity.

Since its original description, many reports have highlighted the variable expression of this condition.<sup>1,2</sup> Cutis marmorata telangiectatica congenita is a relatively frequent feature.<sup>3</sup> It is an autosomal dominant trait with highly variable penetrance and expression. Several isolated cases have been reported. Subsequently, it was reported that some cases of Adams-Oliver syndrome appear to have autosomal recessive inheritance, perhaps with somewhat more severe phenotypic effects.<sup>4</sup> Dilated veins are frequently associated and may be the sole abnormality.<sup>4</sup> Hypoplastic or absent distal phalanges are the most common limb anomalies, but defects range from hypoplastic nails to absent hands or lower legs.<sup>5</sup> Here we report a case of AOS associated with significant cutaneous phlebectasia.

### Case Report

A 40 days old male infant was referred to our clinic at Imam Khomeini hospital of Ahvaz

(Ahvaz Jundishapur University of Medical Sciences, Iran) for cutaneous lesions and poor weight gain. The infant's parents complain poor feeding and vomiting. He was delivered at term by vaginal delivery after a normal pregnancy. Apgar scores were 8 at 1 minute and 9 at 5 minutes. The child was born at 38-week gestational age with a birth body weight of 1700 g, a birth length of 47 cm and a head circumference of 32.5 cm. No resuscitation was required after birth. His 24 years old mother had one previous pregnancy. The first child was healthy. The parents were healthy and cousins. The only drug used during pregnancy was multivitamin. Body weight was 1900 g. The anterior fontanel measured 2.5×2 cm and had a multiple vertex defect (Figure 1). The distal phalanges and nails of both feet were hypoplastic (Figure 2). The fingers of hands were normal. In examination of skin, generalized cutis marmorata was seen (Figure 3). Dilated veins were seen over trunk and head (Figure 4). The patient was formula fed. Abdominal sonogram was negative for hypertrophic pyloric stenosis. Liver and renal function test was normal. Serum electrolyte and arterial blood gases were normal. Echocardiogram was normal. Regular formula was substituted with hydrolyzed formula. Vomiting subsided, but poor feeding and poor weight gain continued. Body weight at 2 months age was 2200 g. Ophthalmologic examination revealed left eye glaucoma.

### Discussion

Adams-Oliver syndrome, which defined by the combination of limb abnormalities and scalp defects, was initially described in 1945 by Adams and Oliver.<sup>6</sup> It is mostly inherited as an autosomal dominant (AD) trait, but also a suggestive autosomal recessive (AR) mode of inheritance and sporadic cases have been described.<sup>7</sup> Our patient had no family history of congenital deformities of the scalp or extremities. The exact pathogenesis of AOS is unknown. Vascular impairment during embryogenesis has been proposed as a possible mechanism by several authors. Hoyme *et al.*<sup>8</sup> reported that the placentas villous vessels of patients with AOS contained multiple organized thrombi. They hypothesized that *in-utero* vascular thrombotic accident led to interruption of blood supply to developing structures. Other reports suggested that AOS is the result of the thrombotic interruption of blood supply in the subclavian, vertebral or other arteries through embryonic period.<sup>9</sup>

Swartz and colleagues suggested that the abnormalities in AOS developed because of a generalized abnormality in small vessels causing disruption of blood flow.<sup>10</sup> Interruption of

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Key words: Adams-Oliver syndrome; absence defect of limbs; Cutis marmorata telangiectatica congenita; glaucoma.

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blood flow through small arteries would account for the aplasia cutis congenita, terminal transverse limb defects, as well as the cardiac, hepatic, and pulmonary vascular lesions. Decreased stability of embryonic blood vessels toward tensile forces during the period of 6 to 8th week of embryonic life due to a gene defect may explain the pathogenesis of vascular anomaly in AOS.<sup>11</sup> Patel and colleagues suggested that abnormal pericyte recruitment to blood vessels may be a possible etiology.<sup>12</sup> The most frequently observed limb malformations in this disorder include syndactyly, brachydactyly, polydactyly, oligodactyly and hypoplastic finger/toenails. There is a great variability in severity of clinical manifestation ranging from the complete absence of the foot or hand to only mild manifestations or normal appearance, as seen in obligate AOS carriers.<sup>13</sup> Aplasia cutis congenita are most frequently found on the vertex of the skull with variable depth and size. Skull defects underlying the scalp lesions may be found.<sup>14</sup> Other associated defects with AOS include cutis marmorata telangiectasia congenita. Cutis marmorata telangiectatica congenita was found in 25% of reported cases and often involve the entire skin including the scalp.<sup>15-17</sup> Ulceration related to particularly large dilated vessels in the skin, as occurred in our patient, is a recognized complication of cutis marmorata telangiectatica.<sup>18</sup> Prominent cutaneous and subcutaneous atrophy and linear depressions overlying dilated vessels on the chest and abdomen has been described previously.<sup>19</sup> Terminal transverse limb defects associated with cutis marmorata telangiectatica without aplasia cutis congenita



Figure 1. Multiple vertex skin defect and dilated veins.



Figure 2. Hypoplastic fingers and nails.



Figure 3. Generalized cutis marmorata.



Figure 4. Dilated veins and skin ulcers.

has been described.<sup>20</sup>

Terminal transverse limb defects significantly affect the distal phalanges or entire digits. Both lower and upper limb defects can be seen, but lower limb defects are more common. Shortening of the fingers with loss of the terminal phalanges of the foot is the most common defect.<sup>4</sup>

## Conclusions

Various expressions of AOS have been reported. This report shows a case of AOS without major multiple organ abnormalities but with poor feeding and abnormal weight gain that may alter the normal lifespan.

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