

Congenital Scalp Aplasia Revealing Adams Olivier Syndrome

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Abstract

Adams-Oliver Syndrome is characterized by the combination of aplasia cutis congenita and limb anomalies. It was initially described in 1945 by Adams and Oliver. Different clinical phenotypes may be related to variable severity both of aplasia cutis and terminal transverse limb defects, and of minor clinical features as cutis marmorata telangiectatica congenita, congenital cardiac defect and vascular anomalies. We, herein, describe a case of 3-month-old girl with aplasia cutis, cutis marmorata telangiectatica and terminal transverse limb reduction defects.

Keywords: Adams-Oliver syndrome; Aplasia cutis congenita; Cutis marmorata; Telangiectatica congenita

1. Introduction

Adams-Oliver syndrome (AOS) is a hereditary polymalformation syndrome currently classified among ectodermal dysplasias. It is characterized by the association of congenital skin aplasia and extremity abnormalities. It is a rare hereditary condition of autosomal dominant inheritance. We report a Moroccan case of an infant presenting the early signs of sporadic type of ODS and fatal outcome.

2. Case report

A 2-month-old girl, Y. Malak, has been brought to the emergency room for management of vertex ulceration since birth. Third child of apparently healthy parents, unrelated, this girl was born vaginally with a good adaptation to extra uterine life. She also had intermittent cyanosis during cries and feedings. The dermatological examination found an oval ulceration of the vertex, purulent bottom, covered with haemorrhagic crusts, 9 cm long and without underlying bone defect (FIG. 1). There

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was also generalized mottling (cutis marmorata) going back to birth (FIG. 2). The rest of the somatic examination also found deformed hands and feet like arthrogryposis (FIG. 3).



FIG. 1. Aplasia cutis congenita on the scalp of our patient.



FIG. 2. Cutis marmorata telangiectatica congenita.



FIG. 3. Arthrogryposis of hands and feet.

Criteria	Clinical feature
Major features	
Aplasia cutis congenita	Limited to scalp vertex may involve dura
Terminal transverse limb defects	
Family history of Adams-Oliver syndrome	Wide phenotypic variability
Minor features	
Cutis marmorata telangiectatica congenita	Livedo reticularis and superficial telangiectasias
Congenital cardiac defect	Atrial septal defect, ventricular septal defect, tetralogy of Fallot, left sided obstructive lesions
Vascular anomaly	Arterial hypoplasia, hepatoportal sclerosis, broncho-pulmonary hemangioma, arterial aplasia

The presence of two major is sufficient for diagnosis while the combination of one major and one minor is denoted a high likelihood of AOS.

FIG. 4. Proposed clinical criteria for diagnosis of Adams-Oliver syndrome.

The rest of the physical examination revealed ectasia of aorta and pulmonary artery, inter ventricular communication and hepatomegaly. Transfontanellar ultrasonography and ophthalmologic examination were normal. The parent's genetic survey did not find any abnormality of the limbs, neither alopecic cicatricial area. The evolution was marked by the installation of a severe bronchoalveolitis, leading to a deterioration of her cardiopathy, then to death.

3. Discussion

AOS is a rare genetic disease, with an estimated incidence of 0.44 per 100,000 live births. The origin of this pathology still unknown until now, although some authors suggest a mechanism of vascular interruption to explain the occurrence of congenital cutaneous aplasia and transverse defects of the limbs [1,2]. The mode of transmission is autosomal dominant with incomplete penetrance, but sporadic or autosomal recessive cases have been reported [2]. Our case is sporadic since the investigations did not reveal similar lesions in ascendants who have no inbreeding link.

The clinical expression is variable, varying from asymptomatic forms to lethal forms with various systemic complications. Congenital dermal aplasia and extremity abnormalities are constantly observed in this syndrome. The congenital cutaneous aplasia mainly concerns the scalp, it is preferentially localized on the vertex [2] and variable in size and depth, sometimes with underlying cranial crest. Manifestations such as syndactyly, polydactyly, oligodactyly, phalanx agenesis and anonychia have been described with variable severity from a person to another [2]. Other manifestations may occur such as cardiac, vascular (cutis marmorata telangiectatica congenita) or cephalic abnormalities (microcephaly, encephalocele).

Specific criteria have been developed by the Algaz team for the diagnosis of AOS described in Board [3,4] (FIG. 4). The diagnosis of AOS is certain when there are two major criteria [5]. Our patient had a complete expression of the disease including two major criteria (aplasia congenital cutaneous, extremity abnormalities) and two minors (cutis marmorata, cardiac abnormalities).

The follow-up of the mother's next pregnancies should focus on the prenatal diagnosis of possible congenital malformations in order to have a prenatal diagnosis [2].

AOS brings together many clinical forms varying from benign malformations to lethal forms. The prognosis depends on the associated forms, mainly cardiac and neurological malformations, which justify systematic screening for appropriate management.

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