 DOI: 10.5578/khj.10430

Iloprost Therapy for Adams Oliver Syndrome With Eisenmenger Syndrome

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ABSTRACT

Adams-Oliver syndrome (AOS) is characterized by scalp defects with terminal transverse limb anomalies. Most reports on this syndrome demonstrate autosomal dominant pedigrees. Cutis marmorata telangiectasia congenita accompanies the syndrome in many cases.In this case we described an AOS with cardiac malformation and eisenmenger syndrome. Since it is a rare case in our literature, the detailed case and literature data are presented.

Keywords: Adams Oliver Syndrome, İloprost Therapy, Eisenmenger Syndrome

Eisenmenger Sendromunun Eşlik Ettiği Adams Oliver Sendromlu Hastalar için İloprost Tedavisi

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ÖZET

Adams-oliver sendromu(AOS) skalp defekti ve uç uzuv anomalisi ile karakterizedir.Birçok rapor bu sendromun otozomal dominant kaltımını kanıtlamıştır.Birçok vakaya kutis marmarotus telenjektazia konjenita eşlik etmektedir.Bu vakada biz AOS'na eşlik eden eisenmenger sendromu ve kardiyak malformasyon tanımladık.Literatürümüzde bulunan çok nadir bir olgu olduğu için detaylı olgu ve literatür bilgisi hazırlandı.

Anahtar Kelimeler: Adams oliver sendromu, iloprost tedavisi, eisenmenger sendromu

Geliş Tarihi: 22.10.2015 - **Kabul Tarihi:** 12.11.2015

INTRODUCTION

Adams-Oliver syndrome (AOS) was first characterized in 1945 by Adams and Oliver, who described eight members of one family in three generations, who had defects of limbs,

scalp and skull [1].

Adams-Oliver syndrome is a rare congenital disorder including congenital scalp and skull defects, extremity anomalies and occasional mental retardation, congenital heart disease [2-5]. The association of cutis marmorata telengiectatica congenita with this syndrome has been described[3,4,6] . The expression of scalp defects sometimes includes bone deformities, and limb defects can vary from nail dystrophy to complete absence of distal extremities. Some cases of Adams-Oliver syndrome occur randomly as the result of a spontaneous genetic change (i.e., new mutation) but AOS is mostly inherited as an autosomal dominant trait but also a suggestive autosomal recessive mode of inheritance has been described[7-8].

CASE REPORT

A 24 years-old girl was admitted to the cardiology unit for dispnea aetiology six years after the establishment of atrial-septal defect.İn physical examination facial dismorphism can be seen, also the other bone structural defects emerge after x-ray examination.In the electrocardiograpy record RBBB with the average heart rate 75/min. ECG revealed right axis deviation and right ventricular hypertrophy.The two dimensional echocardiography

showed an ASD (secundum) with severe pulmonary hypertension & a bi-directional shunt.There was a grade III tricuspit valve insufficiency (Figure 1).Biochemical analysis of blood was within normal limits.

Clinically,she had cyanosis ,clubbing ,cardiomegali, left parasternal heave and epigastric pulsations. The first heart sound was normal and the second was wide and fixed split with accentuation of the pulmonary component. Murmurs of tricuspid and pulmonary regurgitation were also present. The chest examination was clear.

Asymmetric transverse limb reduction defect of right foot, absence of second, third, fourth and fifth phalanx, was observed and hypoplasia of the distal phalanx of the left foot toes and all nails of right and left foot showed partial to complete dystrophy (Figure 2)

Patient had abdominal skin defect with abnormally dilated blood vessels visible directly under the affected skin which is defined as cutis marmorata telangiectetica congenita (Figure 3-4).There were no scalp or limb defects in the family history.

We have followed patient for five years.we treated patient with iloprost,warfain,diüretic and digoxin.Despite patient receiving iloprost for 3 years,we showed no decrease in pulmonary arterial pressure.Although there is no decrease pulmonary arterial pressure,NYHA functional capacity decreased 3 to 2.

DİSCUSSİON

Adams-Oliver syndrome is a congenital condition comprising congenital scalp defects and distal limb abnormalities. A variety of associated features have been reported

including cutis marmorata teleangiectatica, central nervous system abnormalities and cardiac malformations.Variable expression of this syndrome has been reported in

literature[1-5].

Limb defects are typically asymmetric and can be more severe in one arm or leg. The full spectrum of defects that can be observed, ranges from hypoplastic nails, cutaneous syndactyly, transverse reduction defects, polydactyly, and brachydactyly[9]. More severe defects include complete

hand or foot absence, or virtual limb absence[2-3]. This case showed right and left lower limbs involvement.

The patient had congenital heart malformation,namely atrial septal defect and eisenmenger syndrome Zapata et al.[10] analyzing the literature found that 15 out of the total 112 cases of Adams-Oliver syndrome (13.4%) had congenital cardiac malformation.

The range of observed defects included: ventricular septal defect, atrial septal defect, tetralogy of Fallot, coacrtation of the aorta, bicuspid aortic valve, pulmonary

venous stenosis and pulmonary hypertension...

The pathophysiological mechanism of Adams-Oliver syndrome has been a focus of discussion in recent publications... Although a clear pathogenesis has not been yet established, vascular impairments have mainly been assumed. Schwartz et al. hypothesized that a congenital vascular abnormality is the underlying pathogenesis[11]. They suggest that AOS should not be considered a syndrome but rather a constellation of clinical findings resulting from an early embryonic vascular abnormality. With the genetic cause underlying AOS being unknown. Verdyck et al. selected several candidate genes implicated in craniofacial and limb development in order to identify the AOS gene[12].

CONCLUSİONS

Adams Oliver Syndrome may include cardiac anomalies and pulmonary hypertension may rapidly develop patients with AOS.We can begin iloprost in these patients for decreased NYHA functional capacity and mortality.

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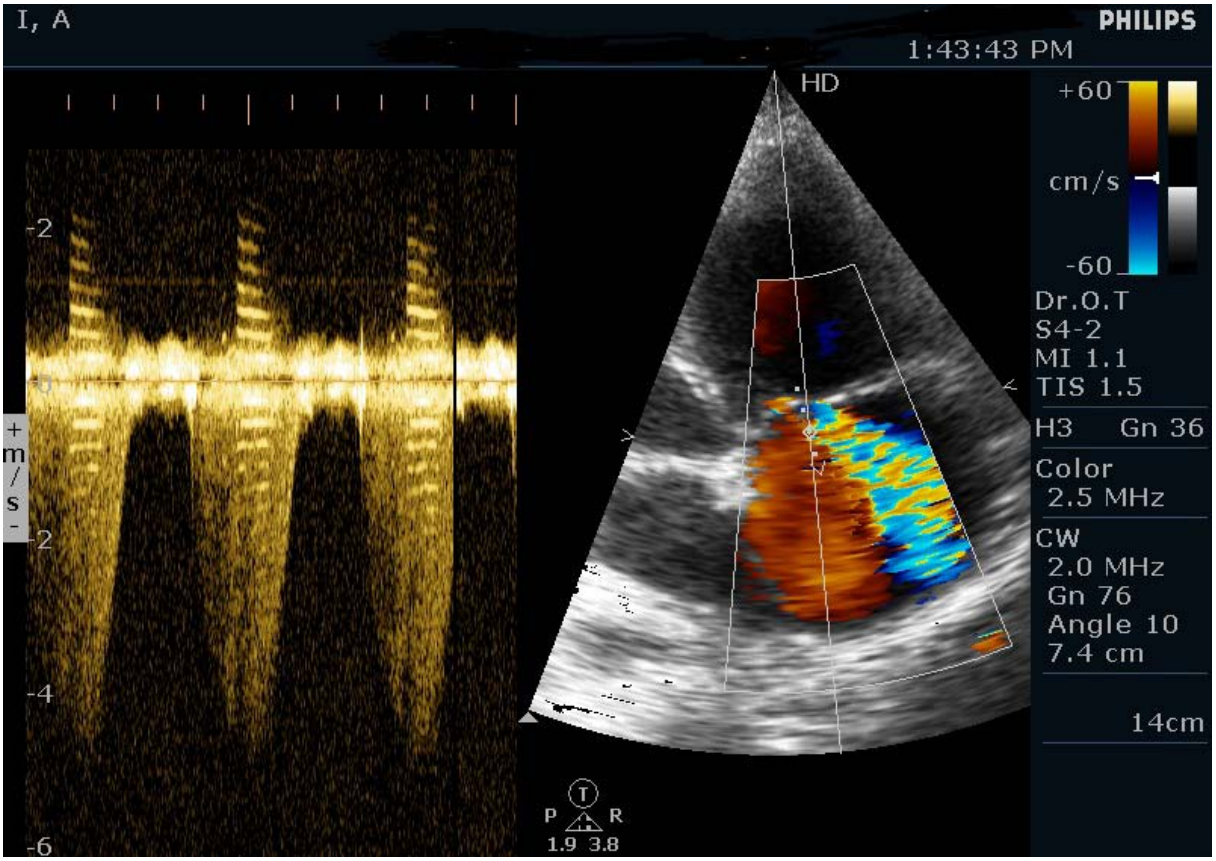

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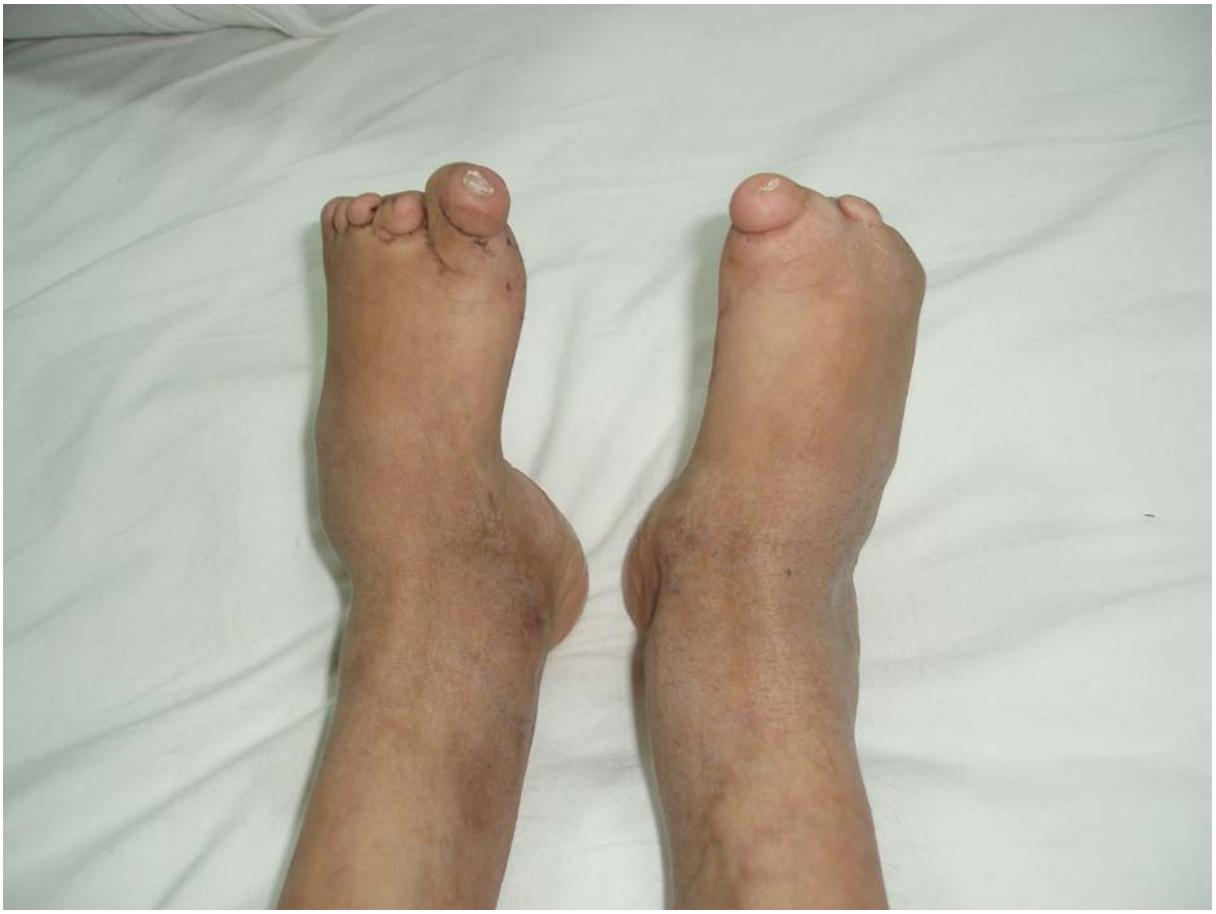

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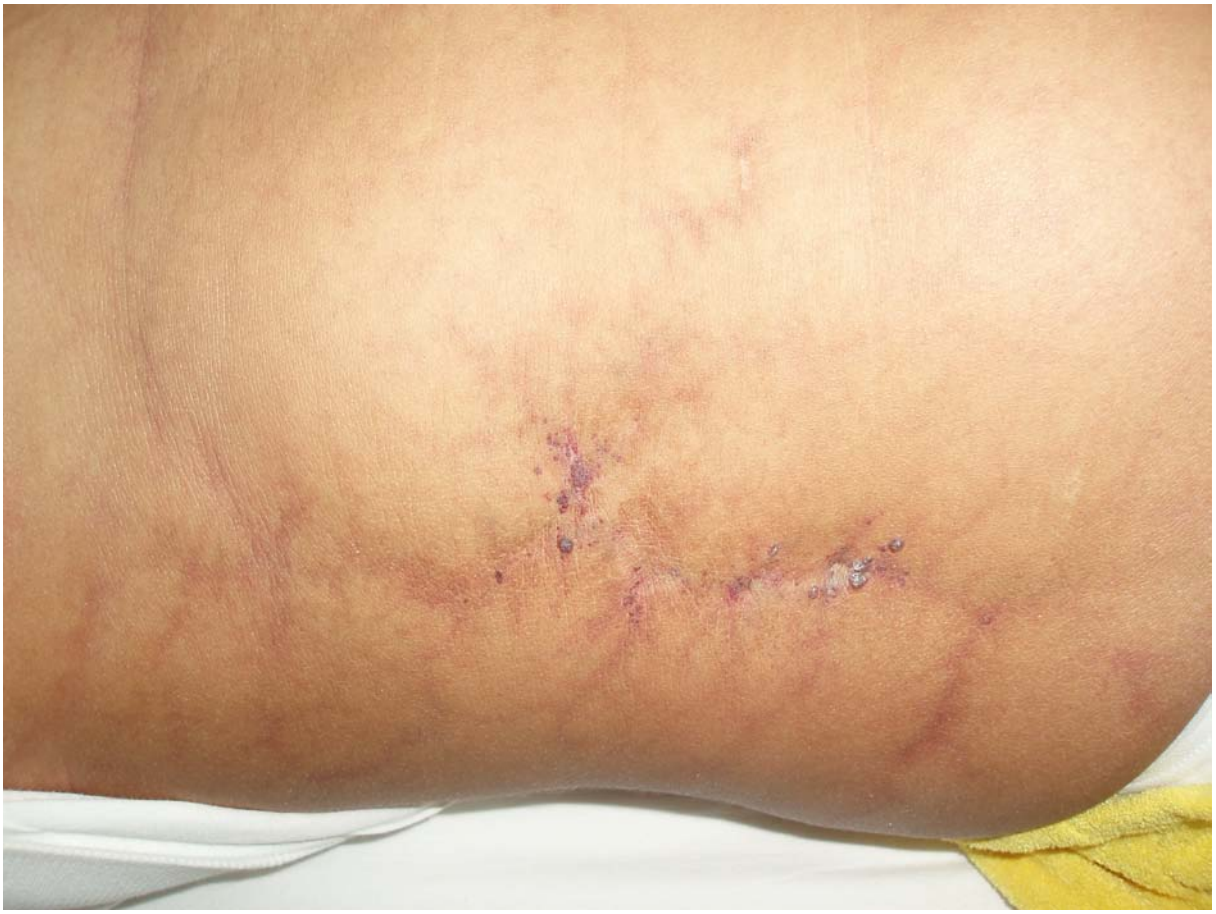

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