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Pulmonary arteriovenous malformation associated with Osler-Weber-Rendu syndrome

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Summary

Osler-Weber-Rendu syndrome is a relatively common vascular displasia characterized by telangiectases of the skin, mucosa, and visceral organs and arteriovenous malformations in children. We reported a 9-year-old boy diagnosed as Osler-Weber Rendu syndrome with his central cyanosis, clubbing, facial and nasal mucosal telangiectases and right pulmonary arteriovenous malformation during an attack of acute rheumatic carditis. After relief of the acute rheumatic carditis attack, his arteriovenous malformation was treated with coil embolization by cardiac catheterization. *(Turk Arch Ped 2011; 46: 256-8)*

Key words: Cyanosis, Osler-Weber-Rendu syndrome, pulmonary arteriovenous malformation, rheumatic heart disease

Introduction

Osler-Weber-Rendu syndrome (hereditery hemorrhagic telangiectasia) is a vascular dysplasia with an autosomal dominant heredity characterized by recurring epistaxis, mucocutaneous telangiectases and arteriovenous malformations (AVM) in internal organs (1). Although the incidence of the disease was reported to be low in the first years when it was firstly defined, it has recently been reported to have an incidence of 1:5000-1:10000 (1-3). Involvement of internal organs include arteriovenous malformations in different numbers in the lung, brain and liver and the incidence of involvement of internal organs in children is not known exactly (4). In this article, a 9 year-old patient who had complaints including epistaxis episodes, getting tired easily and cyanosis in the lips in the last two years and diagnosed as Osler-Weber-Rendu syndrome during an acute rheumatic carditis and whose treatment was adjusted was presented to emphasize the importance of history and physical examination.

Case report

A 9 year-old male patient was referred to us considering acute rheumatic fever, when mitral and aortic failure was found on echocargiography (ECHO) performed in the hospital where he was presented with complaints of fever, weakness and getting tired easily. In the history, it was learned that he had complaints including getting tired easily, cough, cyanosis in the lips and epistaxis episodes especially recurring at night in the last two years. The patient had received treatment for pneumonia after obtaining lung graphy in two centers where he was presented with cough and had had a throat infection with fever one month ago. In familial history, labial bleeding and epistaxis episodes were present.

On physical examination, general state was poor, body temperature was 38,2ºC (axillary), capillary filling time was normal, marked cyanosis was present in the lips and tongue and clubbing was present in both hands. Telangiectases were found on the cheeks and nose. Examination of the circulatory system revealed that apical heart beat was 110/min, blood pressure was 110/40 mmHg and peripheral pulses were natural. While S1 and S2 were heard mildly, S3 was not present. A 2/6 pansystolic murmur was heard on the mitral area and a 2/4 early diastolic murmur was heard in the intercostal space. Examinations of the other systems were found to be normal.

Laboratory findings were as follows: hemoglobin 9.5 g/dL, hematocrite 31%, white blood cells 17 500/mm^3 (neutrophyles

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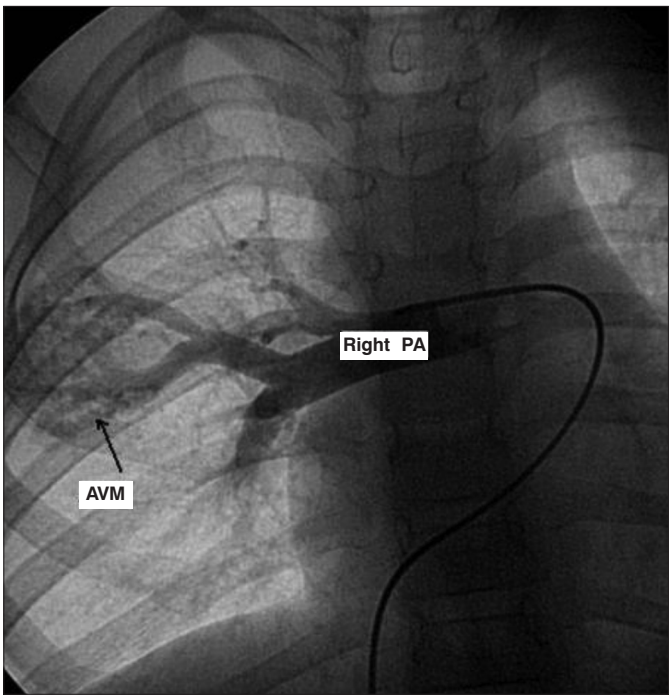
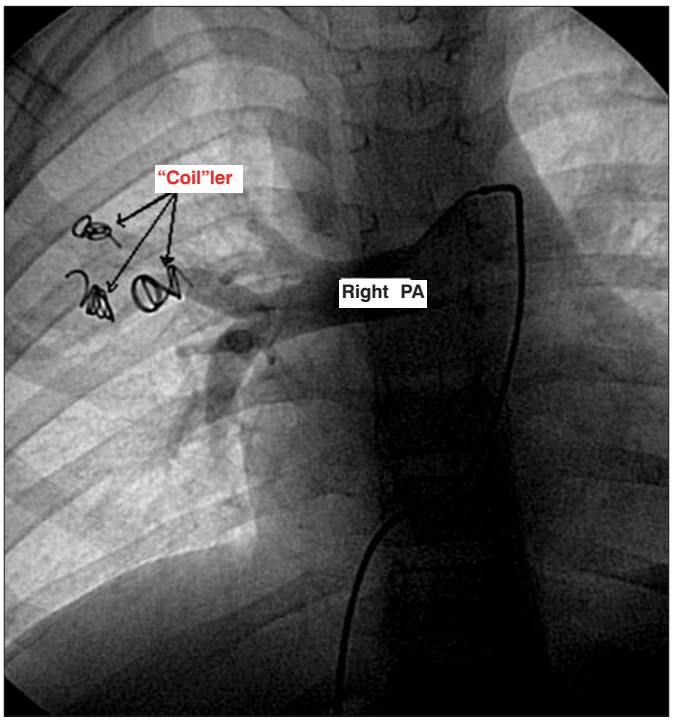
70%, lymphocytes 26%, monocytes 4%), platelet count 513 000/mm^3, C-reactive protein (CRP) 146 mg/L (N<5 mg/L), erythrocyte sedimentation rate 70 mm/hour, anti streptolysine O (ASO) 800 IU. Serum biochemistry tests and throat culture revealed no pathology. Hemoglobin electrophoresis examined for central cyanosis was found to be normal, capillary oxygen saturation was measured to be 88% (in room air) and did not increase by having the patient inhale oxygen. Electrocardiogram was normal and chest graphy revealed mild cardiomegaly (cardiothoracic ratio 56%) and a round opacity with dimensions of 2x4 cm in the right middle zone of the lung. Echocardiogram revelaed significant mitral failure, moderate aortic failure and left atrial and left ventricular enlargement. Pericardial fluid was not observed. A diagnosis of acute rheumatic fever (ARA) was made because of carditis, fever, increase in CRP, erythrocyte sedimentation rate and ASO. Benzathine penicilin, metyl prednisone, digoxin, furosemid and enalapril were started. Although carditis findings and acute phase reactants decreased in follow-up, cyanosis persisted. Pulmonary AVM or fistula were considered in the patient with central cyanosis, telangiectases and findings on chest graphy and contrast ECHO was performed. Following intravenous serum physiologic administration in the right forearm, it was observed firstly the right atrium and right ventricle and in the second cycle the left atrium was filled with bubbles. Lung perfusion scintigraphy revealed findings suggesting intrapulmonary shunt with a rate of 23% and lung tomography with contrast material was taken. On the evaluation of the lung paranchima, an AVM with a nidus of 28x24 mm, an artery of 8 mm and a vein of 12 mm supplied by the right lung upper lobe posterior pulmonary artery branch and draining into the upper lobe posterior pulmonary vein branch was found. A diagnosis of Osler-Weber-Rendu syndrome was made with pulmonary AVM, mucocutenaous telangiectases and recurring epistaxis episodes. No pathology was found on abdominal tomography with contrast material and brain magnetic resonance imaging performed to investigate other possible internal organ involvements. Since the father also had recurring mucocutaneous bleeding episodes, the family was referred to the department of Internal Medicine for screening. Since the patient had active ARF carditis, cardiac catheterization was postponed and the patient was followed up in the outpatient clinic. When 6 months later the findings of ARF carditis were found to be regressed and mild aortic and mitral failure were found on ECHO on the follow-up visit, cardiac catheterization was performed. Selective pulmonary artery angiography performed by entering the right femoral vein revealed an AVM supplied by the right pulmonary artery and draining into the right pulmonary vein (Picture 1). Since the patient was symptomatic and the supplying artery was large, embolization with controlled-released coils with dimensions of 8x5 mm, 6.5x5 mm and 5x4 mm was performed in the supplier arteries of the AVM. After the procedure, control angiography showed that arterial supply of the AVM decreased markedly (Picture 2). Arterial oxygen saturation increased to 95% in the patient whose cyanosis also decreased markedly.



Picture 1. Arteriovenous malformation (AVM) which receives its supplier artery form the right pulmonary artery in the middle zone of the lung on angiography of the right pulmonary artery (PA)



Picture 2. Following "coil" embolization right pulmonary artery (PA) injection for control

Discussion

Osler-Weber-Rendu syndrome is an autosomal dominant vascular dysplasia defined in 1864 for the first time and shown to be familial, to be characterized by telangiectases and to lead mucosal bleeding by Rendu, Osler and Weber, respectively (1-3). The most common pathologic finding is telangiectases and leads to mucocutaneous bleeding. The second most common finding is AVMs observed in the internal organs. The most common regions of involvement are the lung, brain and hepatic circulation (4). The diagnosis is made by showing three or more of the four criteria (epistaxis episodes recurring spontaneously, mucocutaneous telangiectases, AVMs in internal organs (lung, brain, gastrointestinal system) and a diagnosis of Osler-Weber-Rendu syndrome in the first-degree relatives)) (2). It may be difficult to diagnose this disease in the childhood. The reason for this is the fact that mucocutaneous findings are not very prominent or are overlooked in most cases in the first 10 years and recurrent epistaxis episodes which are the most common sign are mostly related to local trauma, upper respiratory infection and allergic diseases in children (3-5). In these cases, if patients with a genetic diagnosis are present in the family, genetic analysis may help to make the diagnosis, since the disease has an autosomal dominant heritance (4). The diagnosis could not be made in our patient, although epistaxis episodes were present for two years before the presentation and telangiectases were found.

Considering all age groups, epistaxis episodes are observed in 90% of the patients and gastrointestinal system bleeding episodes are observed in 10-33% of the patients related to mucosal involvement (3,6). While AVMs in the lung are observed in 15-33% of the patients with internal organ involvement and in the brain in 5-10%, the frequency of AVMs in the liver is not known (1,3,4,7). AVM may be as a single lesion or may be diffuse in the lung. As the age advances, it may grow especially in pregnancy (3,8). Patients with AVM in the lung may be asymptomatic for a long time. In symptomatic patients, respiratory complaints, exercise intolerance and cyanosis are frequently observed (3,5). However, early diagnosis and treatment are important, since sometimes the first sign may be life-threatening lung bleeding, stroke due to right-left shunt or brain abcess (3,4,8). In patients with AVM in the lung, the lifelong incidence of stroke is reported to be 30% and the lifelong incidence of brain abcess is reported to be 5-9%, but this risk seems to be increased in patients with a supplier artery of larger than 3 mm and with multiple supplier arteries (8,9). The diagnosis is made with echocardiography with contrast material, computerized tomography and angiography. In treatment, transcatheter embolization has been used safely and efficiently and decreased the need for surgery. Embolization is recommended in all symptomatic and asymptomatic cases with a supplier artery of larger than 3 mm. With this treatment the rate of right-left shunt is substantially decreased and the risk related to the brain may also be decreased. Lung transplantation can be considered in patients with very diffuse lung AVM (8,10). Our patient was diagnosed during an ARF carditis attack, although he had findings including mucosal bleeding, cyanosis, getting tired easily and respiratory signs for about two years. We could not find an association of ARF and Osler-Weber-Rendu syndrome in the literature. However, we think that this is not a significant association and does not have a cause-effect relation. Since the supplier artery of the AVM in the right lung was 8 mm, transcatheter embolization was performed to prevent complications related to right-left shunt and to decrease cyanosis.

Consequently, although the diagnosis of Osler-Weber-Rendu syndrome is difficult to make in children, a detailed history and physical examination are very important. If no cardiac casue can be found in a patient presenting with central cyanosis, AVM in the lung should absolutely be investigated. In patients in whom AVM is found in the lung, transcatheter embolization should be performed, if the supplier artery is larger than 3 mm to prevent brain complications even if the patient is asymptomatic.

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