A very rare case of polysplenia syndrome with congenital diffuse pulmonary arteriovenous fistulas

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SUMMARY: Gürses D, Ülger Z, Levent E, Özyurek AR. A very rare case of polysplenia syndrome with congenital diffuse pulmonary arteriovenous fistulas. Turk J Pediatr 2006; 48: 96-99.

A five-year-old girl patient was admitted with cyanosis and dyspnea, which started from birth. She had small telangiectatic lesions on her face and cerebral arteriovenous malformation, but no family history of hereditary hemorrhagic telangiectasia. Contrast echocardiography and pulmonary angiography demonstrated diffuse pulmonary arteriovenous fistulas. The patient was diagnosed as polysplenia syndrome characterized with left atrial isomerism, interrupted inferior vena cava, azygous continuation to the superior vena cava, and hepatic veins draining to the right atrium. In contrast to the other polysplenia syndrome cases, in this patient, pulmonary arteriovenous fistulas were not associated with cavopulmonary anastomoses or liver disease.

Key words: pulmonary arteriovenous fistula, polysplenia syndrome, cyanosis, childhood, hereditary hemorrhagic telangiectasia.

Pulmonary arteriovenous fistulas (PAVFs) are abnormal connections between pulmonary arteries and veins. PAVFs are a rare but recognized cause of cyanosis in childhood[1,2], and may be congenital or acquired. Acquired fistulas may occur secondary to trauma such as penetrating chest injury. They may also develop postoperatively in the setting of cavopulmonary connection or Fontan circulation[3,4]. A close relationship between a PAVF and hereditary hemorrhagic telangiectasia (HHT) has often been reported[5]. Development of PAVFs was very rarely reported in cases with polysplenia syndrome after cavopulmonary anastomoses[3,4]. We report here a very unique case with congenital diffuse PAVFs and polysplenia syndrome without intracardiac anomaly, cavopulmonary anastomoses or hemorrhagic telangiectasia.

Case Report

A five-year-old girl was admitted to our hospital with the symptoms of central cyanosis and dyspnea. She had suffered from cyanosis since the newborn period. She was the fourth child of healthy nonconsanguineous parents. The past medical and family histories were unremarkable.

The height was 104 cm (10p) and weight 15 kg (10p), and vital signs were stable. Central cyanosis and digital clubbing were present. The capillary oxygen saturation was 68%. The heart and lung auscultation findings were normal. The liver was palpated on the left side of the abdomen. Neurological examination was normal.

Complete blood count revealed polycythemia. The liver function tests and karyogram were normal. The chest X-ray showed bilateral increased pulmonary vascular markings. Both lungs were morphologically left-sided. On electrocardiographic examination, p wave axis was left superior (–45 degrees) and QRS axis was right inferior (+135 degrees). Arterial blood gas analysis in room air revealed systemic desaturation with partial oxygen saturation value of 40 mmHg and oxygen saturation value of 66%. Methemoglobin level was within normal limits (0.5 g/dl). Transthoracic echocardiography did not reveal any intracardiac pathology but showed direct hepatic vein connections to the right atrium. The contrast echocardiogram following the injection of saline agitated into the peripheral vein showed opacification in the left atrium after two cardiac cycles, thereby suggesting

the presence of intrapulmonary right-to-left shunting (Fig. 1). Abdominal ultrasound examination demonstrated left-sided liver and right-sided polysplenia. Magnetic resonance angiography of the thorax revealed that the vena cava inferior was located on the left side of the aorta, then passed across the vertebra to the right side and continued with azygous and hemiazygous veins. Cranial computerized tomography (CT) revealed an arteriovenous malformation containing multiple telangiectasias. Lung perfusion scintigraphy using ^{99m}Tc showed multiple segmental perfusion defects in the lung and accumulation in the brain and kidneys. The lung ventilation scan revealed no defects.

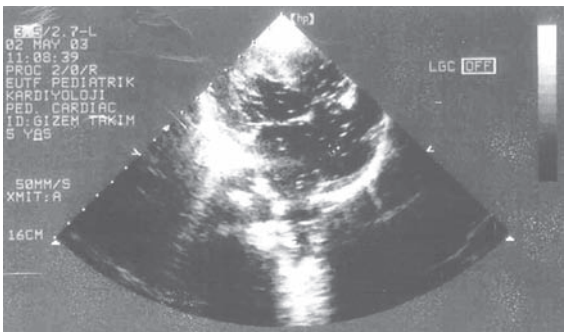

Fig. 1. A "bubble" on contrast echocardiogram (apical four-chamber view). Stream of microbubbles filling the left atrium following systemic venous injection was shown.

The patient underwent cardiac catheterization. The heart was structurally normal, but the inferior vena cava was located on the left side of the aorta, and then passed across the vertebra to the right side and continued with azygous and hemiazygous veins. Pulmonary artery angiography revealed a diffuse fine reticular pattern of pulmonary capillaries on both lung fields (Fig. 2). The pulmonary venous blood was desaturated. These findings were compatible with multiple small arteriovenous fistulas, enabling some degree of bypass of the pulmonary capillary bed. Hemodynamic data obtained at cardiac catheterization are shown in Table I.

Nifedipine was introduced in an attempt to promote pulmonary vasodilatation and thus increase blood flow through physiological capillary channels. The patient was placed on the list for lung transplantation.

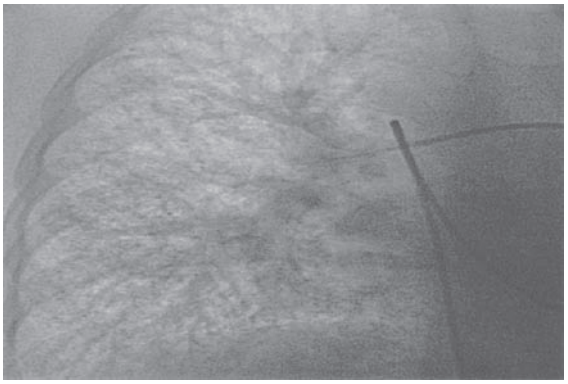

Fig. 2. Right pulmonary artery (PA) angiogram (PA projection) showing a diffuse fine reticular pattern of multiple pulmonary telangiectasias.

Table I. Hemodynamic Data Obtained at Cardiac Catheterization

Site	Saturation (%)	Pressure (mmHg)
Superior vena cava	75	
Right atrium	75	Mean 6
Inferior vena cava	74	
Right ventricle	75	50/5
Main pulmonary artery	75	30/15
Right pulmonary artery	75	30/15
Left pulmonary artery	74	30/15
Left atrium	82	Mean 8
Left ventricle	82	90/5
Aorta	82	90/60

Discussion

Pulmonary arteriovenous fistula has been defined as an abnormal connection between the pulmonary arterial and venous systems that bypasses the pulmonary capillary bed. It may be solitary, multiple, or in the form of diffuse telangiectasia[1,2]. PAVFs are very rare and most often are congenital either as a component of HHT or in an isolated form. Acquired lesions may be associated with liver disease, portal vein thrombosis, trauma, schistosomiasis, or metastatic thyroid carcinoma. They can develop after Glenn shunt or Fontan procedure[3,4]. The pathogenesis of PAVFs remains unclear. Maldistribution of pulmonary blood flow has been suggested as a possible cause of PAVFs[2]. Passive blood return to the lung after a Glenn anastomosis may play a role in the development of PAVFs. Absence of pulsatile pulmonary blood flow has also been implicated in the development of PAVFs.

A close relationship between a PAVF and HHT, also known as Osler-Weber-Rendu syndrome, has often been noted, but the occurrence of PAVFs without a family history of telangiectasia is rare[5]. This syndrome is characterized with recurrent epistaxis, telangiectasias, visceral lesions, and a positive family history[6]. In our patient, there were small arteriovenous malformations in the brain and multiple telangiectasias on the face, but there was no history of epistaxis or family history of HHT. Since the symptoms in HHT are progressive on follow-up, the patient may in future fulfill the criteria of HHT, and thus possibility of HHT could not be completely excluded.

Pulmonary arteriovenous fistulas are a very rare but recognized cause of cyanosis in childhood[1,2,6]. Central cyanosis with the development of finger clubbing may be the first signs indicating the presence of PAVFs. Telangiectasia becomes increasingly prominent, particularly in adulthood. PAVFs are not easily diagnosed routinely, due to their rarity and nonspecific findings on routine examinations. However, this diagnostic hypothesis should always be considered when examining a child with cyanosis, in whom the initial tests do not detect cardiac abnormalities. Two other alternatives in the diagnostic investigation, which have been used in this case, are pulmonary angiography and contrast echocardiography[8].

In our patient, physical examination revealed central cyanosis and clubbing. Electrocardiographic findings were compatible with left atrial isomerism, which was unique for polysplenia syndrome. Contrast echocardiography and pulmonary angiography confirmed the presence of the generalized telangiectatic form of PAVF. Therefore we concluded that in our case, cyanosis was mainly due to PAVFs. Due to the presence of cyanosis since birth and absence of trauma history, PAVFs were thought to be congenital.

Our patient was diagnosed as polysplenia syndrome characterized with left atrial isomerism, interrupted inferior vena cava, azygous continuation to the superior vena cava, and hepatic veins draining to the right atrium. Association of PAVFs with polysplenia syndrome has been very rarely reported[3,4,7]. Amodeo et al.[7] reported two cases with polysplenia syndrome, in whom PAVFs developed after the cavopulmonary anastomoses. They reported that the association of polysplenia syndrome with PAVFs was more than occasional. Srivastava et al.[4] reported 10 patients with PAVFs diagnosed with cardiac catheterization. They had described polysplenia syndrome in six patients with interrupted inferior vena cava and hepatic veins draining to the right atrium. But in all patients, PAVFs developed after the cavopulmonary anastomoses. In only two patients out of 10, PAVFs were unrelated to cavopulmonary anastomoses, and both of these patients had biliary atresia. In contrast to cases from the literature, in our case with polysplenia syndrome, PAVFs were not related with cavopulmonary anastomosis or liver disease.

Polysplenia syndrome may be a predisposition for the development of PAVFs as a part of the incomplete organ development[9]. Cavopulmonary anastomoses or Fontan operation could accelerate this pulmonary complication.

In the patients with unexplained central cyanosis, diffuse PAVFs should be considered. Treatment modalities include both surgical and medical options. Medical treatment is limited to oxygen therapy and pulmonary vasodilators. Nifedipine is one of the few medications available for this purpose. The surgical treatment of PAVFs includes lobectomy, resections and ligation of the fistulas. Transcatheter balloon or coil embolization has proven useful for patients unable to undergo surgical treatment[10]. However, these methods cannot be recommended for patients with diffuse multiple arteriovenous fistulas in the lung, as in the presented case. Lung transplantation seems to be the only way to treat this patient.

REFERENCES

1. Sands A, Daizell E, Craig B, Shields M. Multiple intrapulmonary arteriovenous fistulas in childhood. Pediatr Cardiol 2000; 21: 493-496.
2. Grifka RG, Preminger TJ. Vascular anomalies. In: Allen HD, Gutgesell HP, Clark EB, Driscoll DJ (eds). Heart Disease in Infants, Children, and Adolescents (6th ed). Philadelphia: Williams and Wilkins; 2001: 689-706.
3. Bernstein HS, Ursell PC, Brook MM, Hanley FC, Silverman NH, Bristow J. Fulminant development of pulmonary arteriovenous fistulas in an infant after total cavopulmonary shunt. Pediatr Cardiol 1996; 17: 46-50.
4. Srivastava D, Preminger T, Lock JE, et al. Hepatic venous blood and the development of pulmonary arteriovenous malformations in congenital heart disease. Circulation 1995; 92: 1217-1222.

5. Olgunturk R, Oguz D, Tunaoglu S, İkizler C, Sezgin A, Kula S. Pulmonary arteriovenous fistula in the newborn: a case report of Rendu-Osler-Weber syndrome and a review of the literature. Turk J Pediatr 2001; 43: 332-337.
6. Gianisella RB, Filho RIR, Zielinsky P. Diagnosis and therapeutics of pulmonary arteriovenous fistula in childhood. Case report and review of the literature. Arq Brass Cardiol 2001; 77: 278-281.
7. Amodeo A, Donato R, Carotti A, Marino B, Marcelletti C. Pulmonary arteriovenous fistulas and polysplenia syndrome. Thorac Cardiovasc Surg 1994; 107: 1378-1379.
8. Van Hare GF, Silverman NH. Contrast two-dimensional echocardiography in congenital heart disease: techniques, indications and clinical utility. J Am Coll Cardiol 1989; 21: 255-261.
9. Papagiannis J, Kanter RJ, Effman EL, et al. Polysplenia with pulmonary arteriovenous malformations. Pediatr Cardiol 1993; 14: 127-129.
10. Dutton JA, Jackson JE, Hughes JM, et al. Pulmonary arteriovenous malformations: results of treatment with coil embolization in 53 patients. Am J Roentgenol 1995; 165: 1119-1125.