Pulmonary arteriovenous fistula in the newborn
A case report of Rendu-Osler-Weber syndrome and a review of the literature

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SUMMARY: Olguntürk R, Oğuz D, Tunaoğlu 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.

In most instances, congenital arteriovenous fistula is only one manifestation of a more widespread abnormality; 60% of patients also have hereditary hemorrhagic telengiectasis (Rendu-Osler-Weber syndrome). Among those with congenital pulmonary arteriovenous fistula, the diagnosis is made during infancy in only 15% of patients.

We present a case of pulmonary arteriovenous fistula in a newborn and review the literature. This rare condition of newborns can be treated with different surgical procedures. Only 17 cases of newborn pulmonary arteriovenous fistula/ have been reported, and only two of those had associated Rendu-Osler-Weber syndrome. The results of surgical procedures were good in most of these cases. We treated our case with lobectomy successfully.

Key words: arteriovenous fistula, Rendu-Osler-Weber syndrome.

Pulmonary arteriovenous fistula (PAVF) is one of the rare causes of persistent cyanosis in the newborn. Only 17 cases in the newborn period have been reported to date[1-17]. PAVF is usually associated with Rendu-Osler-Weber (ROW) syndrome in adults and children. However, in newborns only two cases among the 17 were reported to have this syndrome. Most PAVF cases are diagnosed at older ages because it is infrequently considered in the differential diagnosis of neonatal cyanosis and respiratory distress syndrome. The results of surgical repair are usually good even during the neonatal period. We present this rare case with a review of the literature in order to emphasize its diagnostic, clinical and therapeutic features.

Case Report

A 3250 g female baby of 36 weeks' gestation was delivered by cesarean section. Generalized cyanosis was noticed within the first hours, but there were no signs of respiratory distress. The focal dense area at the right upper hemithorax suggested pulmonary arteriovenous malformation, and computerized tomography supported the diagnosis. However, the parents refused further evaluation of the cyanosis, and baby was discharged.

Seven months later, on her second admission, the baby was still deeply cyanotic and without any respiratory problem. Her motor and mental development were normal. The family history was significant with telangiectases in her father, paternal grandfather, and paternal aunt and uncle. Her paternal grandfather had an accessory kidney in the pelvic region. Pedigree of the case is shown in Figure 1. On physical examination: weight was 9500 g, length 75 cm, heart rate 114 beats/min, blood pressure 90/60 mmHg and respiratory rate 62/min. She had generalized cyanosis with minimal clubbing. Pulse oximetry showed O_2 saturation as 65%. There was a 2 x 2 mm telangiectasia on the forehead and 2 x 2 cm hemangioma on the interscapular region. A low-grade continuous murmur was heard at the back on the right side. The liver was palpated 1 cm below the right costal margin. The rest of the physical examination was within normal limits.

Laboratory findings: hemoglobin level was 15.9 g/dl, hematocrit was 47.4%, and methemoglobin level was 1.4% (N: < 15%).

The electrocardiogram and the echocardiogram were in normal limits. Chest X-ray revealed a dense region at the upper and middle lobe of the

right hemithorax. Pulmonary arteriovenous malformation was suspected (Fig. 2), and was supported by thoracal computerized tomography.

The venous and arterial O_2 saturations at the cardiac catheterization were 40% and 59%, respectively. The right heart pressures were within normal limits. A pulmonary arteriovenous fistula involving the upper and middle segments of the right pulmonary artery was seen on the selective posteroanterior cineangiocardiograms (Fig. 3). Nephrogram showed two collecting systems on the right side which abdominal ultrasonography later confirmed to be an extra kidney on the right.

The baby was operated for right upper and middle lobectomy. Postoperative course was uneventful, with no cyanosis or respiratory problems.

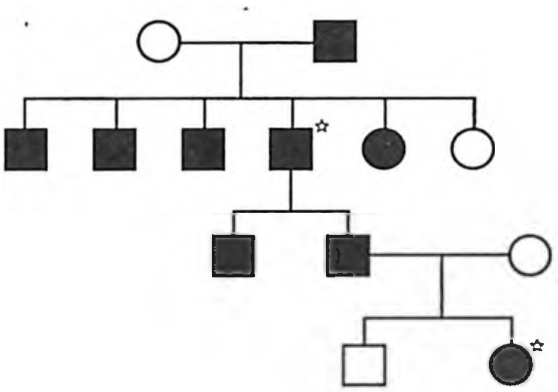

Fig. 1. Pedigree of the case. ☆: Hereditary hemorrhagic telangiectasia (HHT) + kidney anomaly.

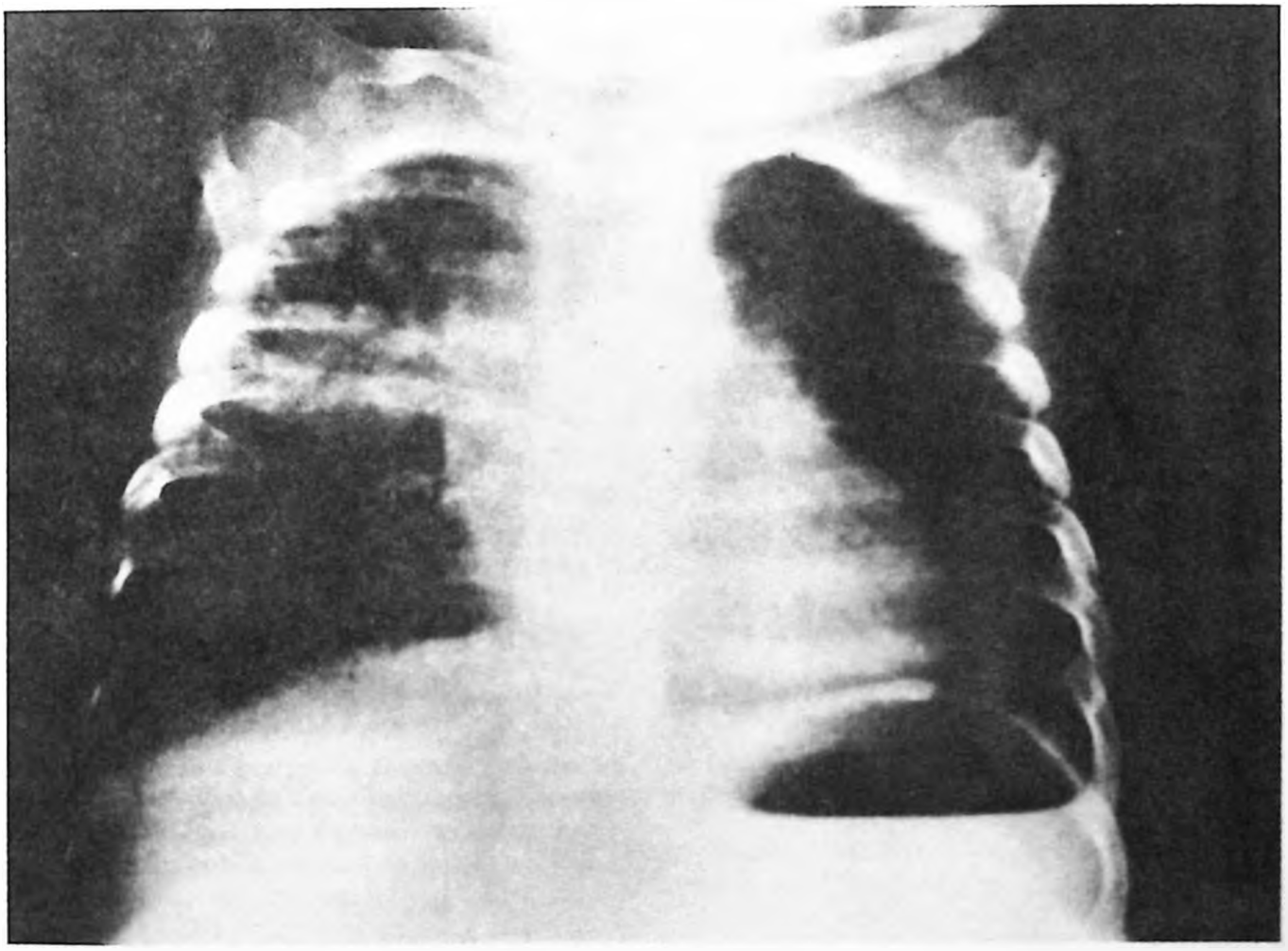

Fig. 2. Chest X-ray.

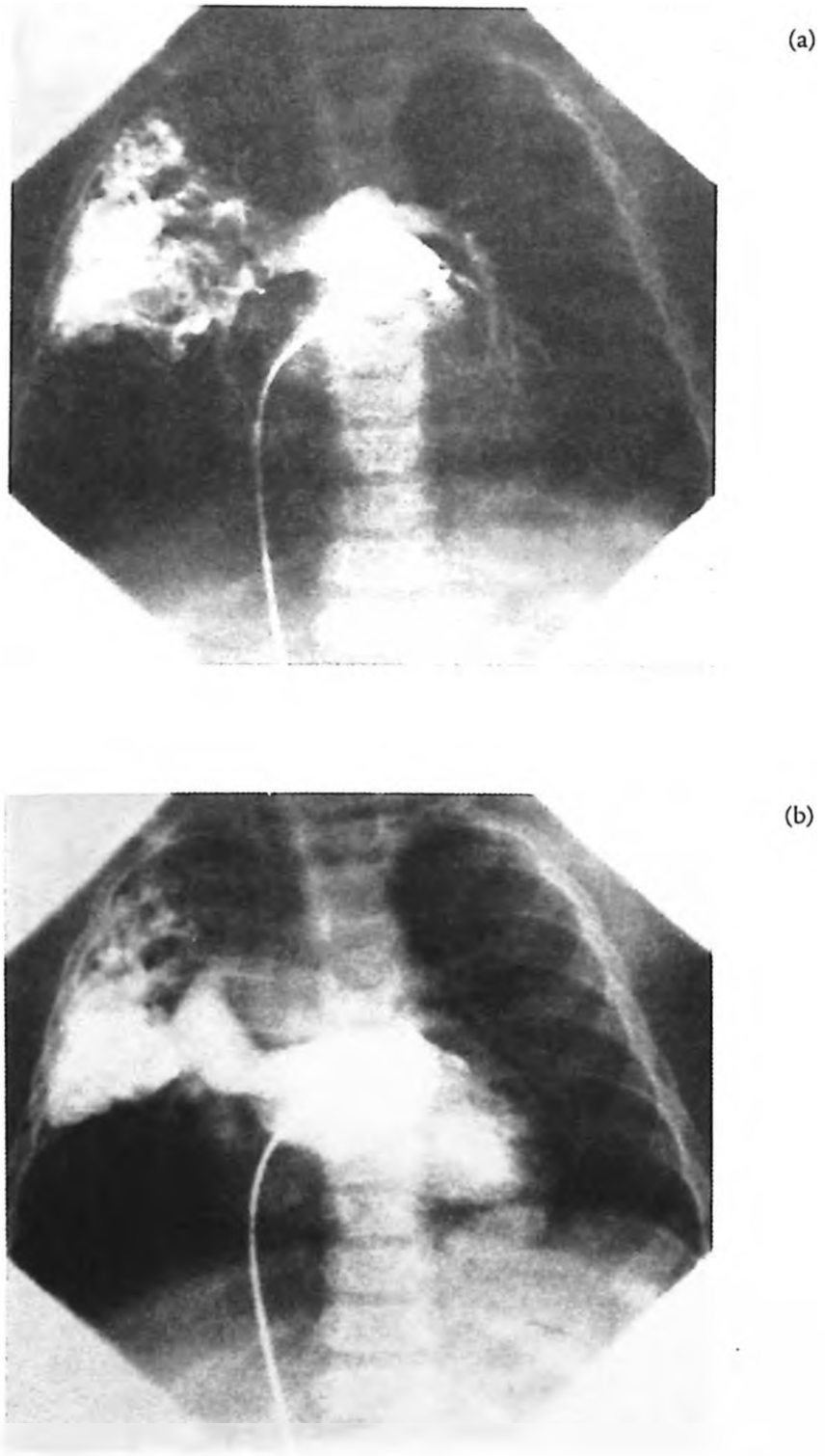

Fig. 3. Pulmonary arteriogram: a) pulmonary arteriogram capillary phase. b) pulmonary arteriogram venous phase.

Discussion

Pulmonary arteriovenous fistula is a congenital malformation of the pulmonary vasculature in which there is persistence of one or more sizable communications that bypass the pulmonary capillary bed, thus diverting unoxygenated pulmonary arterial blood directly into the pulmonary venous system. Familial cases are usually associated with Rendu-Osler-Weber syndrome or hereditary hemorrhagic telangiectasia (HHT), which is an autosomal dominant disorder characterized by multiple mucous membranes and cutoneous telangiectasia, and PAVF's. Previous reports of PAVF have noted the occurrence of HHT in as many as 35-50% of adult cases (Table I)[4-6].

Table I. Summary of the Reported Case of PAVF Presenting in the Newborn

Author Year	Age at diagnosis sex	Cyanosis arterial O_2 % Saturation	Murmur	CHF	Respiratory distress	Diagnosis				Treatment and age	Outcome	+ Anomaly or syndrome
						Chest X-ray	Angiography	CT MRI	Echo USG			
Sweet (17) 1947	1 day Male	Yes	No	No	No	RML	+			Lobectomy 2 years	Good	–
Kafka (1) 1961	2 days Male	Yes	No	No	No	RML	+			Lobectomy 2 years	Good	
Hall (8) 1965	3 days Male	Yes	Yes CM	Yes	No	LLL	+			Resection 6 days	Good	
Crosby (9) 1975	1 day Male	Yes	Yes CM	No	No	LLL	+			Resection 36 hours	Good	PA
Batisse (10) 1977	1 day Male	No	No	Yes	Yes	RML	+			Lobectomy 20 months	Good	–
Batisse (10) 1977	1 day Male	Yes	Yes CM	No	No	LUL	+			Lobectomy 2 months	Good	–
Clarke (11) 1976	1 day Male	Yes	Yes	Yes	Yes	RML	+			Resection	Died	–
Clarke (11) 1976	3 days Male	Yes	Yes	Yes	No	LLL	+			Resection	Good	–
Gula (12) 1981	4 days Male	Yes	Yes	Yes	No	RUL	+			Resection	Good	–
Fried (13) 1982	1 day Female	Yes 27	Yes CM	Yes	Yes	RML+RLL	+	–	+	Death at catheterization	Died	–
Taylor (6) 1983	1 day Male	Yes 30	Yes SM	Yes	Yes	LUL	+	–	+	Death at Surgery	Died	–
Gonzalez (3) 1985	Newborn Female	?	?	No	?	RLL			+	Resection 5 months	Good	ROW?
Vincent (14) 1986	1 day Male	Yes	No	Yes	No	RLL	+			Lobectomy	Good	–
Milovic (15) 1989	2 weeks Female	Yes	No	No	No	LUL	+			Resection	Good	Hemothorax
Allen (4) 1993	1 day Male	Yes 70-80	Yes CM	Yes	No	RLL	+		+	Lobectomy 4 days	Good	ROW
Mitchell (16) 1993	2 weeks Female	Yes 88	Yes SM	No	No	RML	+	–	–	Lobectomy 35 months	Good	
Amaral (2) 1996	6 Days ?	Yes	No	Yes	Yes	LUL			+	Death	Died	?
Olguntürk 1998 (Present Case)	1 day Female	Yes 50	Yes CM	No	No	RUL+RML	+	+	+	Lobectomy 12 months	Good	ROW Accesory kidney

LLL: left lower lobe; RML: right middle lobe; RUL: right upper lobe; RLL: right lower lobe; LUL: left upper lobe; ROW: Rendu-Osler-Weber; PAVF: pulmonary arteriovenous fistula; CHF: congestive heart failure.

Pulmonary arteriovenous fistulas, although recognized as a congenital abnormality, are not often diagnosed until adulthood. Though there are numerous cases of PAVF reported in adults and children, only 17 have been reported in the newborn period (Table I). This is mostly due to the absence of cyanosis in 73% of the newborns affected. Other presenting singns for the newborn are congestive heart failure, dyspnea, bleeding, heart murmur and abnormal chest x-ray. The size of the malformation and the amount of right-to-left flow in these infants determine the symptoms and the time of presentation. Chest X-ray abnormalities occur in 100% of cases. Computerized tomography (CT) and magnetic resonance imaging (MRI) usually confirm the diagnosis[5]. Our case is the only case of a PAVF detected by computerized tomography. A prominent main pulmonary artery, marked pulmonary oligemia and a focal pulmonary density are typical radiological findings[6]. Although noninvasive techniques should be preferred for making the diagnosis, pulmonary artery angiography is still the method of choice. The size and the number of the lesions and the amount of the right-to-left shunt can be determined by cardiac catheterization and angiography. Contrast echocardiography was found successful in determining the diagnosis in one neonate among reported cases[4]. Other echocardiographic clues can be a dilated pulmonary artery, and enlarged left atrium and left ventricle.

Review of te literature as well as our case showed a striking male dominance (12 of 18 cases), in contrast to that seen in older children and adults. Cyanosis was present in almost all cases; murmur and congestive heart failure were the other important symptoms appearing, in 11 and 10 cases, respectively. Respiratory distress was reported in five cases, and four of them died. Respiratory distress as a presenting symptom seems to be a bad prognostic criteria. The location of the lesion, the size of the fistula, and the nature of the rest of the pulmonary arterial bed are the other determinants of survival and of the outcome of surgical correction.

Accessory kidney in our case was probably coincidental. Family history or HHT was mentioned in two previous reports.

Resection of the lesion was performed in all cases and the outcome was satisfactory in 15 cases. Classical treatment in these patients is resection of the lesion or lobe or, infrequently, pneumectomy. Good results have been reported lately with coil embolization, selective miniballoon occlusion and Nd-YAG laser, but these methods are not generally available yet[7]. The risk of systemic embolization also sometimes precludes their use in neonates. The association of Rendu-Osler-Weber syndrome increases the incidence of multiple fistulas. It is important to inform the patient and family of the gastrointestinal, neurologic and genetic aspects of the disease so that appropriate counseling and care may be provided.

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