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A case of prenatally diagnosed extrapulmonary arteriovenous malformation associated with a complex heart defect

Ba-Da Jeong[1], Hye-Sung Won[2], Suah An[2], Ji Yeon Kim[2], Mi-Young Lee[2], Eun Na Kim[3], Jung-Sun Kim[4], Chong Jai Kim[3]

Department of Obstetrics and Gynecology, [1]Ajou University School of Medicine, Suwon, [2]University of Ulsan College of Medicine, Asan Medical Center, Seoul; Department of Pathology, [3]University of Ulsan College of Medicine, Asan Medical Center, Seoul, [4]Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea

Pulmonary arteriovenous malformations are rare vascular anomalies of the lung, only a few cases of which have been diagnosed prenatally. The diagnostic clue for prenatal diagnosis was cardiomegaly with a particularly enlarged left atrium. All previous cases of pulmonary arteriovenous malformations diagnosed prenatally have been reported as an isolated anomaly or in association with simple heart defects. We here describe the first case of a pulmonary arteriovenous malformation with a complex heart defect that was diagnosed prenatally at 21.0 weeks of gestation and confirmed by postmortem autopsy.

Keywords: Congenital heart defects; Prenatal diagnosis; Pulmonary arteriovenous fistulas

Introduction

Pulmonary arteriovenous malformations (PAVMs) are abnormal communications between the pulmonary arterial and venous system. They are quite rare and only a few reports of prenatal diagnosis have been described [1-9]. The cases diagnosed prenatally were reported as an isolated anomaly or in association with simple cardiac defects. Here, we present the first case of prenatally diagnosed PAVM associated with double outlet right ventricle (DORV) with ventricular septal defect (VSD) that was confirmed by autopsy.

Case report

A 32-year-old primigravida woman was referred to our center at 21.0 weeks of gestation with suspicion of fetal DORV with VSD. The non-invasive prenatal test was normal. The ultrasound examination showed cardiomegaly (cardiothoracic area ratio, 0.47; normal range, 0.25 to 35) with a particularly enlarged left atrium (Fig. 1A). Both great arteries arose from the right ventricle with a dextro-transposition relationship, and a huge VSD (6.5 mm) was present (Fig. 1B). The aortic and ductal arches were observed on the left side of trachea. However, the main pulmonary artery was widened (Z-score 2.19) and coursed abnormally to the right side of the trachea, turned to the left behind the lower level of the trachea, and joined to the left atrium. Pulsed wave color Doppler was applied to the area of the junction of the pulmonary artery and left atrium and showed an arterial flow pattern, which confirmed it as a right-to-left shunt with suspicion of a PAVM (Fig. 1C). The pulmonary valve appeared normal without thickening and laminar antegrade flow with normal velocity was demonstrated at color and pulsed Doppler. The blood flow through the ductus arteriosus was reversed. The baby was delivered due

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Corresponding author: Hye-Sung Won
Department of Obstetrics and Gynecology, University of Ulsan College of Medicine, Asan Medical Center, 88 Olympic-ro 43-gil, Songpa-gu, Seoul 05505, Korea
Tel: +82-2-3010-3630 Fax: +82-2-3010-6944
E-mail: hswon@amc.seoul.kr
http://orcid.org/0000-0003-0611-2401

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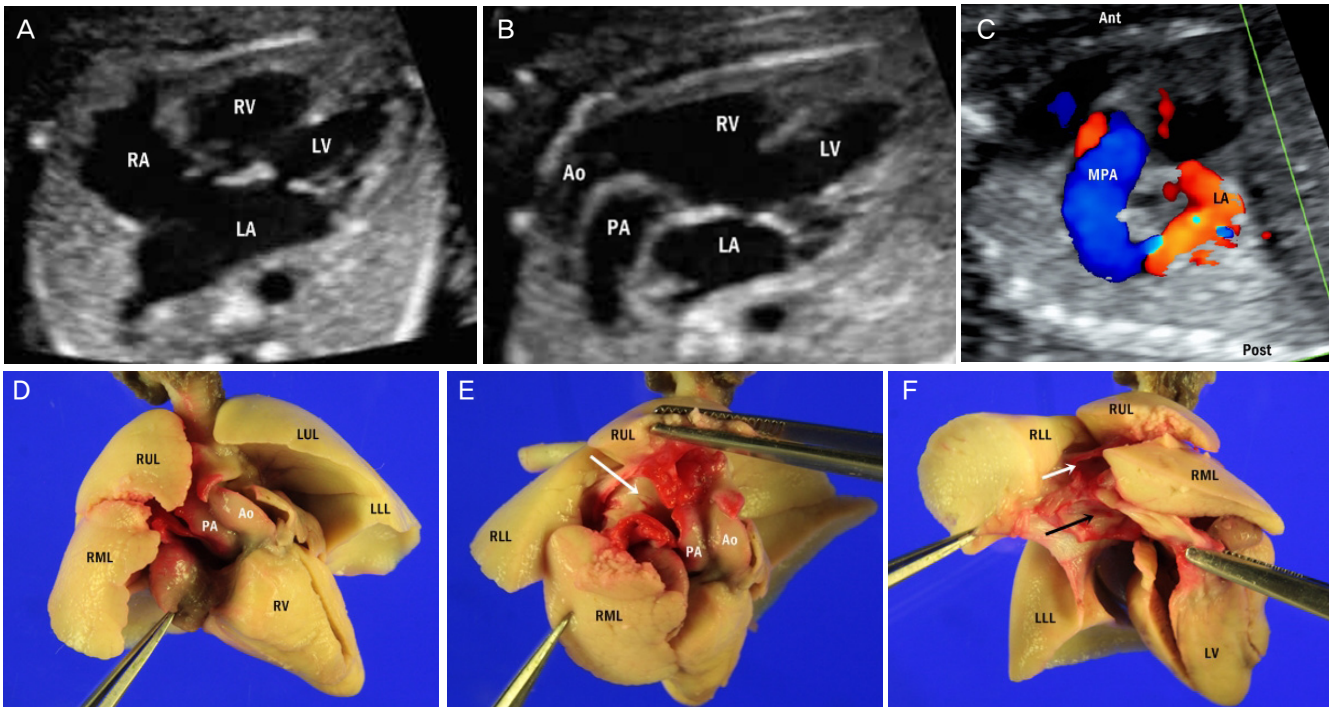

Fig. 1. Fetal echocardiographic images obtained at 21.0 weeks of gestation in the current study case demonstrating cardiomegaly with a particularly enlarged left atrium (LA) in the four-chamber view (A), both aorta (Ao) and pulmonary artery (PA) arising from the right ventricle (RV) (B), and an abnormal communication of main pulmonary artery (MPA) and LA with color Doppler in the parasagittal view (C). Postmortem autopsy showing the abnormal root of Ao in front of the pulmonary artery (PA) arising from the RV in the anterior view (D), PA from the RV running between the right upper lobe (RUL) and the right middle lobe (RML) of the lung turning to the left behind the main bronchus in the right superior view (white arrow, opened PA) (E), and PA connecting to the LA in the right posterior view (white arrow, opened PA; black arrow, opened LA) (F). The inside of LA was dyed by red ink flowing through the PA. RA, right atrium; LV, left ventricle; Ant, anterior; Post, posterior; LUL, left upper lobe; LLL, left lower lobe; RLL, right lower lobe.

to impending fetal death but expired. The autopsy confirmed the extrapulmonary PAVM between the main pulmonary artery and the left atrium, which was associated with DORV and VSD (Fig. 1D-F). The histological investigation could not differentiate between the pulmonary artery and vein due to the small size of the heart. There was no evidence of telangiectasia or further malformations.

Discussion

PAVMs result from persistent capillary anastomoses or defects in the terminal capillary loops allowing dilatation and formation of vascular sacs [10,11]. The incidence of this disease is 2 to 3 per 100,000 people, and more than 80% of PAVMs are congenital [5,12]. They may be an isolated anomaly or associated with hereditary hemorrhagic telangiectasia in about 70% of cases [2]. The clinical symptoms of patients are variable from asymptomatic to dyspnea depending on the degree of right-to-left shunt through PAVMs [13]. Patients with small lesions remain asymptomatic and may not present until the fourth decade of life [2]. The frequent complications are cerebrovascular attacks or brain abscesses by emboli entering the systemic circulation in the absence of the pulmonary capillary filter [14].

Technological advancements of ultrasound such as color and pulsed wave Doppler help an earlier prenatal diagnosis of congenital abnormalities. Similar with previous reported cases, PAVMs could be diagnosed prenatally by ultrasonographic findings of cardiomegaly particularly left atrium, abnormal course of pulmonary artery and pulsed Doppler of arterial drainage into left atrium. In addition to these findings, it is the first case of a PAVM associated with a complex heart defect diagnosed prenatally. In previous cases of PAVM diagnosed prenatally, three were associated with a congenital heart defect, but a simple disease such as valvular pulmonary stenosis,

Table 1. Previous reported cases of fetal PAVM diagnosed prenatally

Reference	Center	GA at diagnosis (wk)	Site of PAVM	Associated anomaly	GA at delivery (wk)	Fetal hydrops	Perinatal outcomes
Kalugdan et al. (1989) [8]	Fukuoka, Japan	28	In RUL RPA to RPV		31	+	Expired after 4 hours before treatment
Heling et al. (2002) [3]	Berlin, Germany	27	In RUL, RLL RPA to RPV		30	+	Expired during transcatheter occlusion
Russell et al. (2002) [6]	Michigan, USA	23	In RLL RPA to RPV	Valvular PS	NA	-	EXIT, ECMO, surgical ligation → expired due to pulmonary hypoplasia
Kenny et al. (2007) [4]	Bristol, UK	37	In RML, RLL RPA to RPV		Term	NA	Transcatheter occlusion at HD 1 → survived
Sinkovskaya et al. (2009) [7]	Virginia, USA	22	RPA to RPV		39	-	Transcatheter occlusion at HD 1 → survived
Akler et al. (2012) [1]	Tel Aviv, Israel	16	MPA to RPV	Muscular VSD	-	NA	Termination of pregnancy
Hellmund et al. (2014) [9]	Bonn, Germany	22	In RLL RPA to RPV	ASD, muscular VSD	36	-	Surgical closure at HD 1 → survived
Ostras et al. (2015) [5]	Kyiv, Ukraine	21	LPA to LPV		38	-	Surgical repair at HD 2 → survived
Current study	Seoul, Korea	21	MPA to LPV	DORV with VSD	-	-	Preterm delivery due to impending fetal death but expired

PAVM, pulmonary arteriovenous malformation; GA, gestational age; RUL, right upper lobe; RPA, right pulmonary artery; RPV, right pulmonary vein; RLL, right lower lobe; PS, pulmonary stenosis; NA, not available; EXIT, ex utero intrapartum treatment; ECMO, extracorporeal membrane oxygenation; RML, right middle lobe; HD, hospital day; MPA, main pulmonary artery; VSD, ventricular septal defect; ASD, atrial septal defect; LPA, left pulmonary artery; LPV, left pulmonary vein; DORV, double outlet right ventricle.

muscular VSD, and atrial septal defect [1,6,9]. In our case, a complex heart defect of DORV with VSD might add the hemodynamic effect to the already exiting shunting through a PAVM. Several studies have reported that PAVMs occurred after a palliative operation with cavopulmonary anastomoses in patients with a congenital heart defect [15]. However, they were not congenital, but acquired conditions.

In eight previous cases of prenatally diagnosed PAVMs, four infants survived after successful treatment (Table 1) [1,3-9]. Among four survived infants, one was diagnosed as late as 37 weeks. Although the other three fetuses were diagnosed early at 21 to 22 weeks, none of them had signs of heart failure during pregnancy, and all were born at or near term. This may mean that the degree of shunting was insufficient to cause heart failure prenatally.

In contrast to the small PAVMs, large PAVMs might cause a hyperdynamic circulatory status leading to heart failure, even in the antenatal period. As a result, preterm delivery was needed and infants could not survive even after an immediate treatment postnatally [3,6]. The earliest diagnosed case showed a PAVM between the main pulmonary artery and the right pulmonary vein. Similarly, this case was confirmed as an extrapulmonary PAVM with direct connection between main pulmonary artery and the left atrium. It might make the larger amount of shunting than the other seven cases with localized communication between either the right or left pulmonary artery and pulmonary vein inside the lung. As a result, the larger hemodynamic change would occur.

It is difficult to evaluate the prognostic factors of PAVMs diagnosed prenatally because of its rarity. However, this case was suggested as the worst condition showing the hemodynamic change prenatally, the extrapulmonary connection of main pulmonary artery and the additional complex heart defect. In cases of large PAVMs like this, antenatal intervention such as coiling of shunt before fetal heart failure or irreversible pulmonary hypoplasia may be helpful for improvement of

the perinatal outcome in future.

PAVMs are rare, but can be diagnosed prenatally with typical ultrasonographic findings in cases with large hemodynamic change. It may be an isolated form or associated with congenital heart defect ranging variously from simple to complex.

Conflict of interest

No potential conflict of interest relevant to this article was reported.

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