Pulmonary atresia with a ventricular septal defect and left pulmonary artery discontinuity: A case report

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Abstract

Unilateral pulmonary artery discontinuity (UPAD) is a rare malformation which is associated with other intracardiac abnormalities. Cases accompanied by other cardiac abnormalities are often missed on prenatal echocardiography. We reported our case which was prenatally diagnosed as pulmonary atresia with ventricular septal defect (PAVSD) and left pulmonary artery (LPA) discontinuity.

Pulmonary atresia with a ventricular septal defect and left pulmonary artery discontinuity: a case prenatally diagnosed and confirmed by a postnatal computed tomography

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Unilateral pulmonary artery discontinuity (UPAD) is a rare malformation which is associated with other intracardiac abnormalities. Cases accompanied by other cardiac abnormalities are often missed on prenatal echocardiography. We reported our case which was prenatally diagnosed as pulmonary atresia with ventricular septal defect (PAVSD) and left pulmonary artery (LPA) discontinuity.

Key words

Foetal echocardiography, Pulmonary artery, Unilateral pulmonary artery discontinuity

Key Clinical Message

The left and right pulmonary arteries do not always originate from the same great artery (MPA or common arterial trunk); therefore, clinicians should check the route of each pulmonary artery.

Case presentation

A 33-year-old pregnant woman (G1P0) was referred to our hospital at 24 weeks of gestation because of a suspected foetal heart anomaly on routine obstetric ultrasonography. The results of her prenatal laboratory tests were normal. Foetal echocardiography revealed a large ventricular septal defect (VSD) measuring 5 mm with a large overriding aorta (Figure 1A). We also observed multiple major aortopulmonary collateral arteries (Figure 1B). Therefore, the initial prenatal diagnosis was pulmonary atresia with VSD (PAVSD). A very atretic main pulmonary artery (MPA) can be seen in figure 1a retrospectively however; we were not able to detect this artery at that time. The subsequent foetal echocardiography which were performed at 26 weeks of gestation revealed a highly atretic MPA from the right ventricle (RV) giving rise to the right pulmonary artery (RPA), without bifurcation (Figure 2A). Instead of the bifurcation of the MPA, the left pulmonary artery (LPA) originated from the left subclavian artery (LSA; Figure 2B and C). The echogenicity of the thymus was not definitive on prenatal echocardiography. On the basis of these findings, the foetus was diagnosed as having PAVSD with left PAD and 22q 11.2 deletion syndrome. Considering the gestational age at diagnosis, we decided to postpone the genetic study after birth. A female neonate was delivered by elective caesarean section at $37^{6/7}$ weeks of gestation for the timed delivery, with a body weight of 2,740 g, Apgar score of 8/9 points, heartbeat of 155 beats per minute, respiratory rate of 44 breaths per minute, blood pressure of 71/38 mmHg, and SpO_2 of 88%. Multidetector computed tomography (MDCT) revealed a right-sided aortic arch, with the left-sided ductus arteriosus (DA) originating from the LSA and MAPCA. It also revealed a narrow RPA (2.7 mm) connecting with the MPA (2.7 mm), without connection with the LPA (2.7 mm size). The LPA originated from the left-sided DA originating from the LSA. Three-dimensional MDCT images showed the posterior aspect of the heart of the affected neonate (Figure 3A and 3B).

The neonate was assisted using the non-invasive continuous positive airway pressure (CPAP) mode because of chest retraction and tachycardia. For maintaining the patency of the DA, administration of prostaglandin E1 a-cyclodextrin clathrate (PGE_1-CD; Eglandin) 5 ng/(kg·min) was initiated immediately after birth, and the dosage was adjusted between 3 and 5 ng/(kg·min), targeting 85% of the SpO_2. Meanwhile, postnatal multiplex ligation-dependent probe amplification (MLPA) revealed that the neonate's condition was complicated by 22q11.2 deletion syndrome.

The general condition of the neonate remained stable; therefore, we attempted to wean the neonate from CPAP to high flow nasal cannula (3 L) and tapered the PGE_1-CD dosage to 1 ng/(kg·min). However, on the 15th day after birth, the SpO_2fluctuated and required assist control mandatory ventilation in the intubation mode. After stabilisation of the neonatal status, right ventricular outflow tract (RVOT) reconstruction and pulmonary artery re-implantation were performed on the 25th day after birth. Postoperative echocardiography revealed a RVOT without turbulent flow and bilateral pulmonary arteries measuring 4.2 mm (left) and 3.3 mm (right). However, the SpO_2 fluctuated and haemoptysis requiring full sedation occurred on the seventh day after the operation. The subsequent echocardiography revealed decreased blood flow in the right ventricle and RPA with pericardial effusion; thus, right heart failure was suspected. We started milrinone administration to augment ventricular contractility and decrease the afterload. However, the neonate expired because of right heart failure.

Discussion

UPAD is rare congenital malformation and usually demonstrated as an absence of proximal unilateral pulmonary artery on prenatal echocardiography; therefore, it has also been referred to as unilateral absence of the pulmonary artery.[1] As the distal segment of the affected pulmonary artery actually exists, UPAD is a more accurate term to describe this malformation.[1] It also should be distinguished from the other rare form of CHD, hemitruncus arteriosus, which is defined as an anomalous origin of one of the branch pulmonary arteries (PAs) from the aorta and a normal origin of other PAs from the RVOT.[2] If untreated, it results in a large left-to-right shunt, with the whole cardiac output from the right ventricle directed to the unaffected lung while the other lung receives blood at a systemic pressure from the systemic aorta; its 1-year survival rate has been reported to be <30%.[2,3]

Unlike patients with hemitruncus arteriosus, those with UPAD receive blood supply to one lung from ductus-like collateral vessels, not the systemic aorta, and it is often associated with other CHDs, especially tetralogy of Fallot (TOF).[2] Although, its pathophysiology is not fully understood most studies on UPAD have described that a failure in the connection of the sixth aortic arch with the pulmonary trunk results in this developmental anomaly.[1,4] Clinical presentations vary depending on the affected site.[1,4,5] In case of LPAD, 75% of patients have an associated congenital heart disease, including TOF, right-side aortic arch, septal defect, or persistent DA.[1,5,6] By contrast, most patients with RPA discontinuity have the isolated form without other intracardiac anatomies. The affected pulmonary artery is on the side opposite the aortic arch.[6] As known previously, in this case, the LPA was affected, and the neonate showed a right aortic arch. This implies that the confirmation of the aortic arch location has an important clinical significance.

As previous mentioned, the initial diagnosis was PAVSD only. In the case of PAVSD, the initial maintenance of DA is also important because the pulmonary circulation is dependent on DA. However, the atretic MPA was not dependent on DA and was responsible for the right lung circulation in the present case. If PGE1 was administered for a certain period to maintain DA without prenatal detection of UPAD in our case, it would be associated with a decrease in the size of the right PA, resulting in PA size discrepancy. Meanwhile, an isolated form of UPAD (usually affecting the right PA) could be missed during prenatal care. Diagnosis is often delayed in patients with pulmonary hypertension, recurrent pulmonary infections, congestive heart failure, and haemoptysis.[4] Prenatal detection is important because it aids in the prompt initiation of PGE1 administration to ensure early rehabilitation of the affected lung.[7]

No consensus has been reached regarding the treatment for UPAD. In cases complicated by other CHDs, treatment would depend on major cardiac abnormalities. In our case, the prenatal detection of UPAD in addition to PAVSD allowed the paediatric cardiologist to make a precise operative plan. Our paediatric cardiologist knew the size difference between the LPA and the MPA, which made it possible to prepare the Gore-Tex graft patch for pulmonary angioplasty. In cases of the isolated form of UPAD, early intervention for UPAD has been supported owing to the concern for regression of the affected PA after DA closure. However, the operation timing should be determined on the basis of many other clinical situations, including the neonatal condition or birthweight.[7] If early intervention is unavailable, administration of PGE1 is usually required. In cases of delayed diagnosis made in the adolescent or adult period, lobectomy and ligation of the affected PA are required.[5] In conclusion, UPAD is relatively rare, but when undetected, it could affect neonatal prognosis. Therefore, clinicians should examine the route of the both pulmonary arteries regardless of the existence of other intracardiac abnormalities.

Conflicts of interest

The authors declare no conflicts of interest.

Author Contributions

HHC and WJS wrote the paper. HMK, HMK, MJK and WJS: treated the patient

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Figure L egends

Figure 1. Prenatal echocardiogram showing a large ventricular septal defect and an overriding aorta (arrow) and an atretic main pulmonary artery (arrowhead) (A) and multiple aortopulmonary collateral arteries (B).

Figure 2. Prenatal echocardiogram showing an atretic main pulmonary artery giving rise to the right pulmonary artery (arrow) (A), left subclavian artery (LSA) from aortic arch (B), and left pulmonary artery (LPA) arising from the left subclavian artery (C).

Figure 3. Postnatal multidetector computed tomography image showing the posterior aspect of the neonate. Also shown are the right pulmonary artery (white arrow) arising from the atretic main pulmonary artery (A) and the left pulmonary artery (white arrow) arising from the left subclavian artery (arrowhead) (B).