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A case report of prenatal diagnostics and surgical correction of pulmonary agenesis associated with total anomalous pulmonary venous connection and vascular ring

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Abstract
Pulmonary agenesis is a rare malformation associated with congenital heart disease and may lead to a life-threatening condition and death without pre-planned surgical and medical support.
We report an unusual case of unilateral pulmonary agenesis in combination with total anomalous pulmonary venous connection, complete vascular ring, retroaortic, retroesophageal innominate vein, secondary atrial septal defect, which was prenatally identified by fetal ultrasound and magnetic resonance imaging, later confirmed by postnatal computed tomography.

Keywords
Pulmonary agenesis, total anomalous pulmonary venous connection, vascular ring, congenital cardiac malformation

Introduction

Pulmonary agenesis is rare congenital pathology (1 per 15,000 live births) that is commonly associated with other congenital cardiovascular anomalies and syndromes, such as atrial septal defects, ventricular septal defects, heart valve malformations, Shone's syndrome, tetralogy of Fallot, and total anomalous pulmonary venous connection (TAPVC) [1].

The clinical features of unilateral pulmonary agenesis, in the absence of other malformations, are compatible with normal life [2], but there is a high risk of severe respiratory infection during infancy which may lead to pneumonia and death.

Case Report

A 31-year-old woman was present at our center for consultation concerning congenital heart disease in the fetus which was prenatally examined, at term 32 weeks gestation, by echocardiography (Figure 1) and magnetic resonance imaging (MRI) (Figure 1). Heart anomaly was confirmed with the next anatomical findings: left pulmonary agenesis, supracardiac TAPVC, and complete vascular ring made by a right aortic arch with retroesophagal left subclavian artery and left patent ductus arteriosus (with the right to left shunt); this vascular ring encircled and compressed the esophagus along with the trachea. The parenchyma of the right lung was well-developed, according to gestational age. The right pulmonary veins flowed through the vertical vein (passed behind the left atrium) and drained into the retroaortic, retroesophageal innominate vein.

The woman was given the recommendation to plan the childbirth next to our center, as we expected the severe condition of the infant.

A 39-week-gestation female infant was born from a healthy mother with high prenatal risk factors. The Apgar score was 4/5 at 1 and 5 minutes respectively, and the infant weighed 2.58 kg. Meconium was reported in the amniotic fluid. Cyanosis with severe respiratory distress syndrome (FiO2 40%, SatO2 -72%) was also noted. The infant was intubated and urgently delivered to our department where she underwent examination by two-dimensional color echocardiography and CT-angiography. The congenital heart disease and anomalies of the respiratory system were confirmed. The computed tomography (CT) showed the left pulmonary agenesis with left rudimentary bronchus and no vascular supply (type 2 by Boyden) and herniation of the right lung to the left side; the trachea shifted to the left side and was significantly compressed (up to 70%) by the vascular ring. Uneven pneumatization of the lung parenchyma and interstitial emphysema were also noted. The main pulmonary artery was normal size with the hypoplastic right pulmonary artery (diameter up to 3 mm) and diminutive right pulmonary veins (diameter -1,0 mm) (Figure 2).

Open heart surgery (on the 8th day of life) performed by median sternotomy confirmed the anatomy malformation found by echocardiography and CT (Figure 3). Intraoperative bronchoscopy revealed the vascular ring compression of the trachea. It was not possible to pass beyond the narrowing of the trachea to assess the bronchus.

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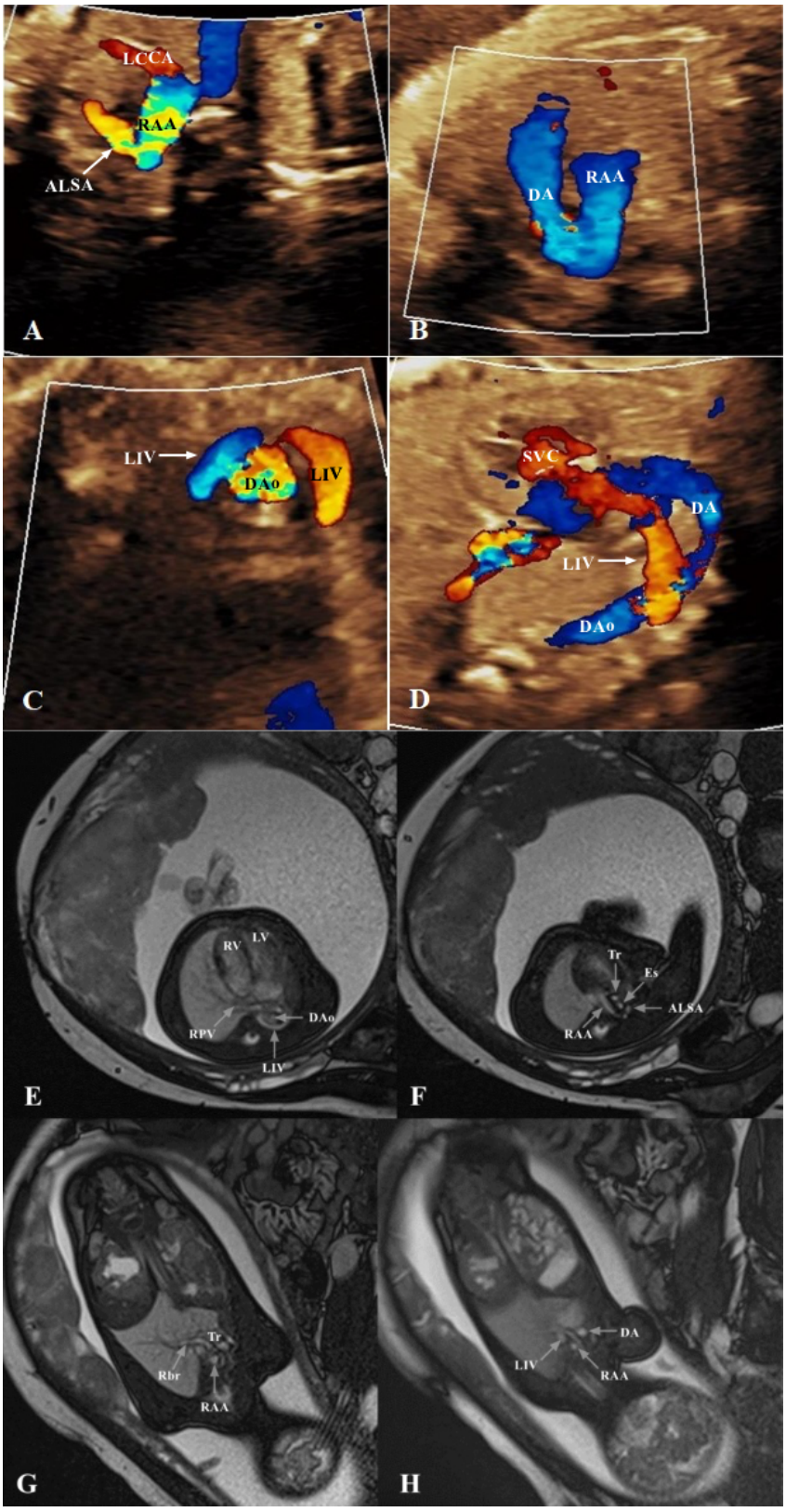


Figure 1 - Fetal echocardiography at 32 weeks gestation shows a complete vascular ring made by a right aortic arch, left ductus arteriosus with the aberrant left subclavian artery (A, B). Retroaortic, retroesophageal innominate vein drained into superior vena cava (C, D). Fetal MRI at 32 weeks gestation showed left pulmonary agenesis, the right pulmonary veins drained into the retroaortic, retroesophageal innominate vein (E, F), complete vascular ring made by a right aortic arch with retroesophagal left subclavian artery, which encircles the esophagus and trachea (G, H).
LIV – left innominate vein; DAo – descending aorta; RAA – right aortic arch; ALSA – aberrant left subclavian artery; DA – ductus arteriosus; SVC – superior vena cava; RV – right ventricle; LV – left ventricle; RPV – right pulmonary veins; Tr – trachea; Es – esophagus; RAA – right aortic arch; Rbr – right bronchus.

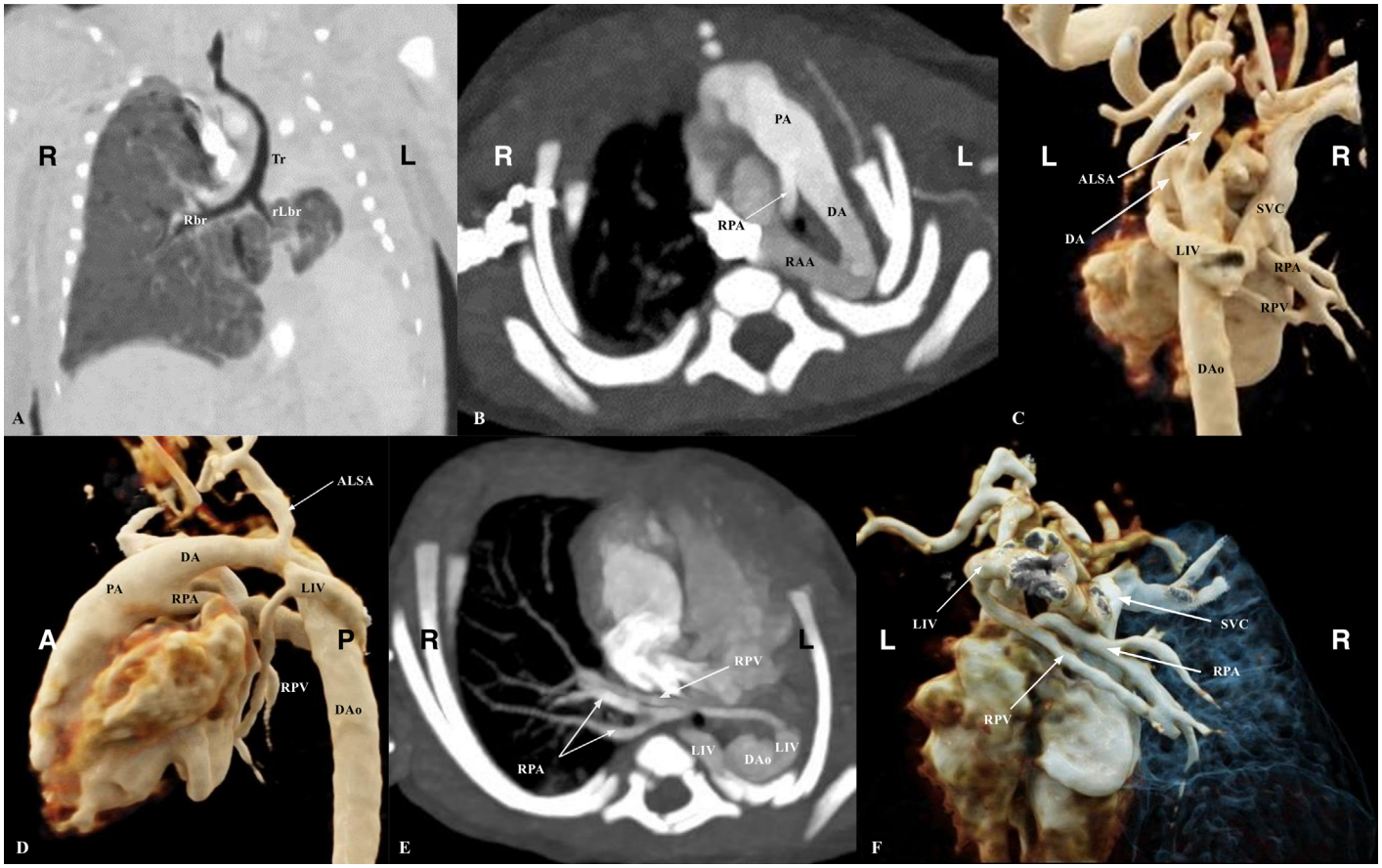


Figure 2 - Postnatal CT shows the left pulmonary agenesis with left rudimentary bronchus (A – minimum intensity projection (MinIP)), trachea shifted to the left side and compressed by the vascular ring, hypoplastic right pulmonary artery (B – maximum intensity projection (MIP)), diminutive right pulmonary veins drained into the retroaortic, retroesophageal innominate vein (C, D, F – 3D volume rendering technique (VRT) and E – MIP).
Tr – trachea; Rbr – main right bronchus; rLbr – rudimentary left bronchus; PA – pulmonary artery; RAA – right aortic arch; DA – ductus arteriosus; ALSA – aberrant left subclavian artery; LIV – left innominate vein; SVC – superior vena cava; RPV – right pulmonary veins; DAo – descending aorta; Es – esophagus; RPA – right pulmonary artery; R – right; L – left; A – anterior; P – posterior.

Vascular ring compression was relieved by dividing the patent ductus arteriosus. Cardiopulmonary Bypass (CPB) was set up, antegrade cold cardioplegia was obtained. The left atrium (LA) and the pulmonary venous confluence (PVC) were anastomosed. CPB stopped, was identified the bleeding from the posterior surface of LA and the sign of pulmonary edema. We decided to reconstruct the anastomosis between LA and PVC, when deep hypothermic circulatory arrest (17° C) started. Specific anatomical features of both right pulmonary veins (1.0 mm diameters) were a predictor of the primary complication during the surgical correction. Several trials to stop cardiopulmonary bypass with maximal medical support including inotropic and cardiopulmonary resuscitation were effectless, and the patient died.

Discussion

Pulmonary agenesis is a rare malformation resulting from abnormal embryonic development, which can be isolated lesions or associated with other anomalies. There are three types of pulmonary agenesis described by Boyden [3]:

- Type 1 (agenesis) – Complete absence of the lung and bronchus and no vascular supply to the affected side;
- Type 2 (aplasia) – Rudimentary bronchus with the complete absence of pulmonary parenchyma;
- Type 3 (hypoplasia) – Presence of variable amounts of the bronchial tree, pulmonary parenchyma, and supporting vasculature.

In the literature review [4—8], we have found just a few references about the combination of unilateral pulmonary agenesis with TAPVC and atrial septal defect, but there was no reported case of unilateral pulmonary agenesis in combination with TAPVC drained into the retroaortic, retroesophageal innominate vein, complete vascular ring, and secondary atrial septal defect. Morbidity and mortality in unilateral pulmonary agenesis are related to associated anomalies, as well as respiratory disease and combination with heart malformation.

Therefore, we should mention that a meticulous prenatal diagnostic is highly important in cases where we suspect congenital lung agenesis, as it is commonly associated with congenital heart disease and may lead to a life-threatening condition and

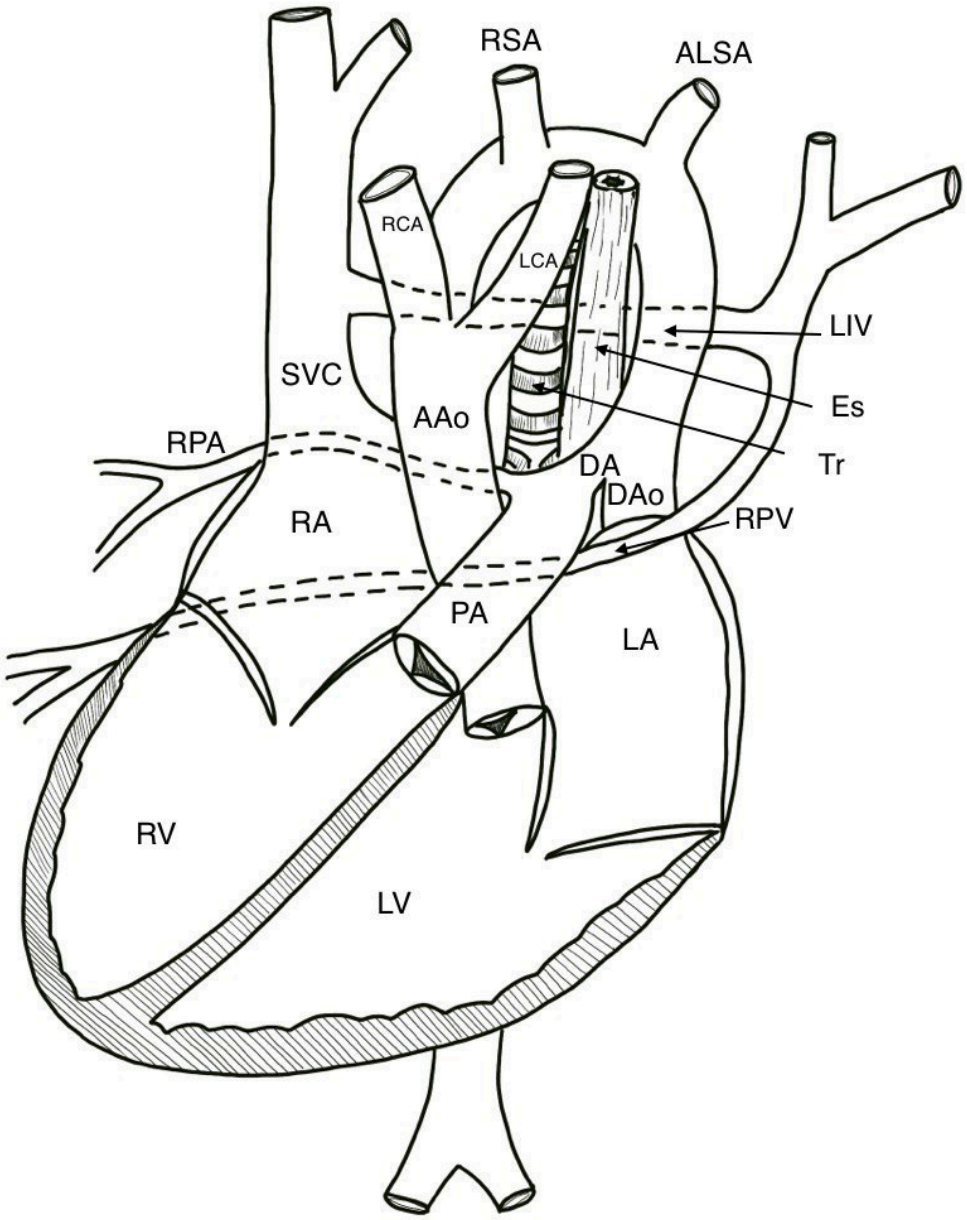


Figure 3 - The diagram shows the anatomy of the heart malformation confirmed intraoperatively.
Tr – trachea; Es – esophagus; PA – pulmonary artery; DA – ductus arteriosus; ALSA – aberrant left subclavian artery; LIV – left innominate vein; SVC – superior vena cava; RPV – right pulmonary veins; RPA – right pulmonary artery; RA – right atrium; RV – right ventricle; LA – left atrium; LV – left ventricle; RSA – right subclavian artery; RCA – right common carotid artery; LCA – left common carotid artery; DAo – descending aorta; AAo – ascending aorta.
The figure was drawn by B. Hulitskyi.

death without pre-planned surgical and medical treatment. A lot of thoracic anomalies have a similar echogenic appearance and because of that our recommendation for prenatal diagnosis of congenital malformations is a combination of two-dimensional color echocardiography and MRI [9], as the latter can overcome the diagnostic difficulties and provide more detailed information about extra lesions and anatomic configuration to reveal the underlying risk factors for surgical correction.

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Conflicts of Interest

None.

Ethical Standards

This manuscript does not involve human or animal experimentation.

Patient Permission

The patient's family gave consent to publish this case report.

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