Cardiology in the Young

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A rare prenatal diagnosis: congenital absence of aortic valve

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Brief Report

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Abstract

Congenital absence of the aortic valve is characterised by the absence of aortic valve and severe regurgitation. The rest of the reported cases were mostly diagnosed either on postnatal echocardiography or autopsy. Here, we present a foetal case with the absence of the aortic valve and "inverse circulatory shunt".

Congenital absence of the aortic valve is a rare pathology reported mostly as single case reports in the literature. Unlike the congenital absence of the pulmonary valve, the congenital absence of the aortic valve is mostly fatal, resulting in hydrops, foetal demise, or early neonatal death.[1,2] The absence of the aortic valve is mainly characterised by the absence of aortic valve structure and free regurgitation of the valve, decreased left ventricular functions, a malignant circulation called an inverse circulatory shunt in which severe mitral regurgitation is directed to the right atrium through foramen ovale. We want to demonstrate the images of this rare pathology with the inverse circulatory shunt.

Case presentation

28-year-old nulligravida was evaluated at 30th gestational week. Foetal echocardiography revealed concordant viscera-atrial situs, D-looped ventricles, normally aligned major arteries, and cardiomegaly. The left ventricle was hypoplastic and did not create the apex; the right heart structures were dilated (Fig 1a and b). Mitral annulus was 5.8 mm (z score: −3.36); tricuspid annulus was 9.7 mm (z score: −0.27). The mitral valve was hypoplastic with severe regurgitation and without significant antegrade flow (Fig 1c). There was a massive left to right shunt from the foramen ovale (Video 1). The aortic valve was dysplastic without any visible valve leaflets (Fig. 1d). Severe aortic regurgitation with the minimal antegrade flow and ascending aorta filling by retrograde ductal flow were observed (Fig 2, Video 2). There was a decrease in a wave velocity of ductus venosus flow, and umbilical artery diastolic flow was also decreased. This severe CHD caused severe heart failure; pericardial, pleural effusions, and ascites were developed, and the fetus died in utero at 33rd gestational week.

Discussion

Congenital absence of the aortic valve is a rare anomaly with high mortality. Although mostly fatal cases with hydrops were presented in the literature, rare cases reported by Harada, Krasemann, and Qasim et al, who were born alive without hydrops and operated, were also reported.[1,3,4] Usually, two different haemodynamic presentations with different clinical outcomes were reported about the congenital absence of the aortic valve.[2,5] One of them is associated with mitral regurgitation and non-restrictive foramen ovale, which generally causes severe hydrops and intrauterine foetal death. Mitral regurgitation causes "inverse circulatory shunt," that is, the regurgitation from the ascending aorta to the left ventricle – left atrium by mitral regurgitation – right atrium through the foramen ovale – right ventricle – pulmonary artery – ductus arteriosus and ascending aorta again.[6] The other presentation is the absence of the aortic valve with mitral atresia or restrictive foramen ovale, which plays a role in blocking the "inverse circulatory shunt" and the hydrops that enable the fetus to reach the term.[7,8] But in our case, the fetus with absence of the aortic valve and hypoplastic left ventricle developed hydrops due to the circular shunt consisting of the aortic regurgitation, severe mitral regurgitation, and large left to right shunt through foramen ovale. This made us think that mitral regurgitation can be more essential in the foetal malignant circulation than the morphology of the left ventricle.

In conclusion, the degree of aortic and mitral regurgitation and the flow through the foramen ovale and the ventricular functions evaluated with foetal echocardiography seems valuable for predicting pre- and postnatal outcomes and counselling patient's pregnancy outcomes. These echocardiographic evaluations are also necessary for planning potential intrauterine treatment, the timing of birth, and postnatal interventions.

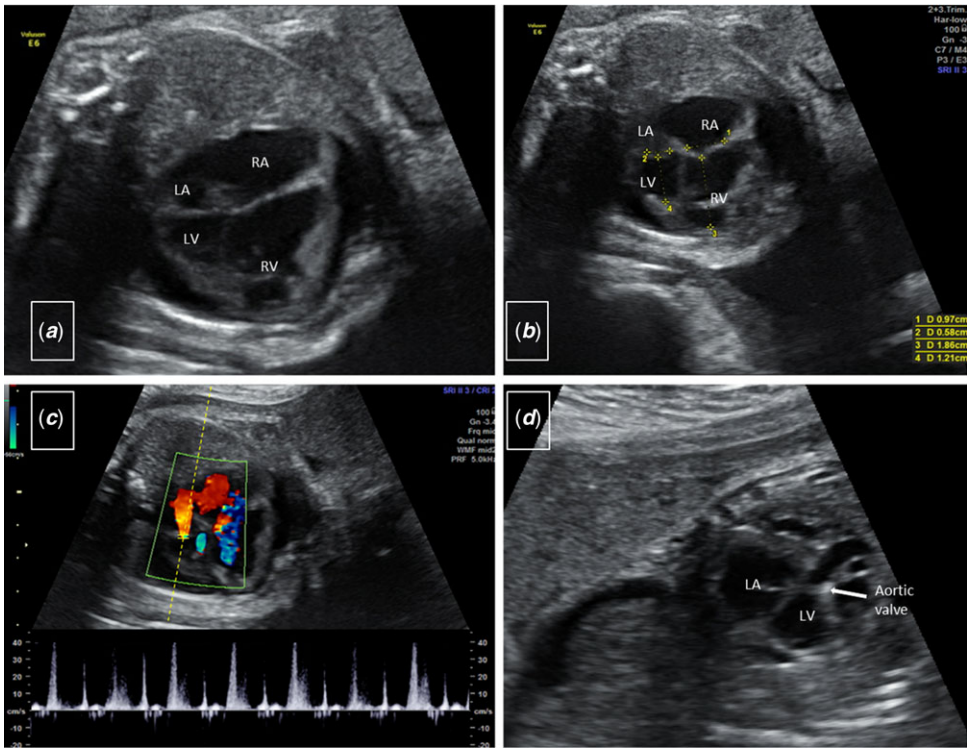

Figure 1. (***a***) Hypoplastic left heart structures with a non-apex forming left ventricle and global aneurysmatic dilation at the right heart structures at four-chamber view. (***b***) Mitral and tricuspid valves and ventricular diameters. (***c***) Severe mitral regurgitation without significant antegrade flow with PW Doppler examination. (***d***) Dysplastic aortic valve without any clearly visible valve leaflets at the annulus. FO = foramen ovale; LA = left atrium; LV = left ventricle; RA = right atrium; RV = right ventricle.

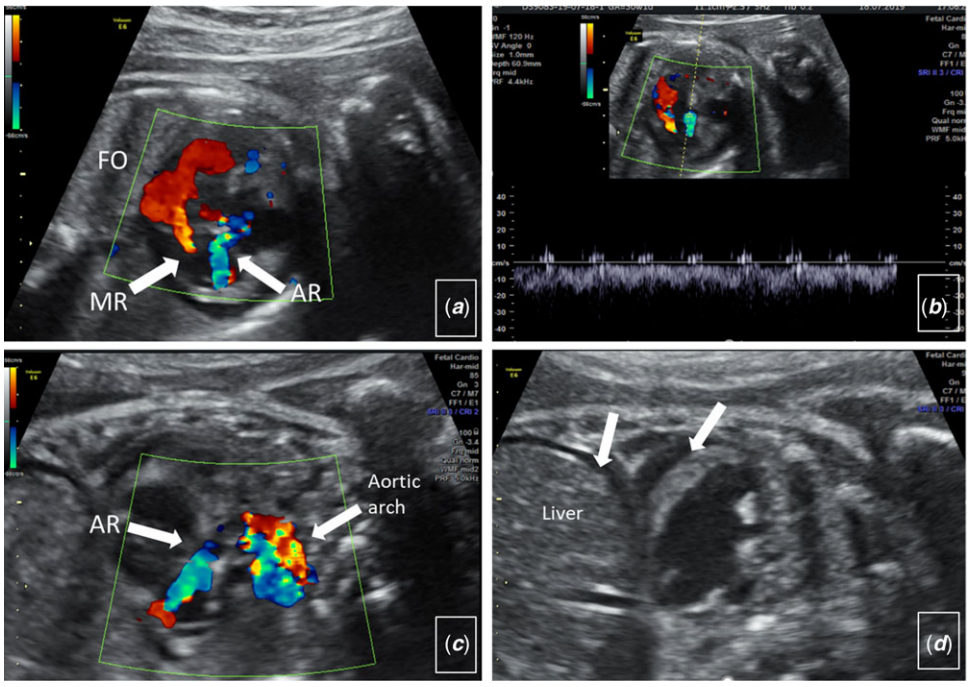

Figure 2. (***a***) Severe regurgitation from the hypoplastic mitral valve continues with a large left to right shunt from the foramen ovale; and severe regurgitation from the aortic valve. (***b***) Severe aortic regurgitation and a bidirectional flow pattern, with the diastolic retrograde flow and limited systolic antegrade flow with PW Doppler examination. (***c***) Severe aortic regurgitation, ascending aorta and aortic arch filling by retrograde ductus flow, red flow in the aortic arch. (***d***) The arrows pointing pericardial effusion and ascites. AR = aortic regurgitation; FO = foramen ovale; MR = mitral regurgitation.

Supplementary material. To view supplementary material for this article, please visit https://doi.org/10.1017/S1047951122000919

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Ethical standards. The authors assert that all procedures contributing to this work comply with the ethical standards of the relevant national guidelines on human experimentation and with the Helsinki Declaration of 1975, as revised in 2008, and have been approved by the institutional committees.

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