Hypoplastic left heart syndrome with endomyocardial fibroelastosis diagnosed in first trimester: A case report

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Case report

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Abstract

Background Report a case of hypoplastic left heart syndrome (HLHS) with endomyocardial fibroelastosis (EFE) diagnosed in the first trimester.

Case presentation We detected a balloon shaped left ventricle, a narrowed reverse blood stream signal in the aortic arch and widened blood stream signals in the pulmonary artery (2.2 mm) and the ductus arteriosus when performing echocardiographic screen with transabdominal sonography in a 31-year-old pregnant woman (gravida 2, para 1) who presented to our department for routine fetal ultrasonography at 11^{+2} weeks' gestation. Based on these findings, a diagnosis of HLHS with EFE was suspected. The findings from autopsy confirmed the diagnosis.

Conclusion Ultrasound screening in the first trimester may be helpful in early diagnosis of HLHS.

Background

Hypoplastic left heart syndrome (HLHS) is a congenital cardiovascular anomaly characterized by hypoplasia of mitral valve, left ventricle, aortic valve and ascending aorta. The incidence of HLHS in live births is 12/100,000, which accounts for 1%-2% of all congenital heart diseases [1–2]. HLHS is usually complicated by endomyocardial fibroelastosis (EFE) when the hypoplasia of mitral valve is severe and/or the aortic valve is severely stenosed or occluded. HLHS can be diagnosed in early pregnancy [3–4]. However, HLHS with EFE diagnosed in first trimester is rarely reported. Here we reported a case of HLHS with EFE diagnosed in early pregnancy.

Case Presentation

A 31-year-old pregnant woman, gravida 2, para 1, with no significant medical history and no family history of congenital heart defects presented to our department for routine fetal ultrasonography at 11^{+2} weeks' gestation. Ultrasound screening revealed a crown-rump length (CRL) of 51 mm and a nuchal translucency thickness of 10.5 mm. Meanwhile, a reversed A wave in the ductus venosus and fetal hydrops was observed. Therefore, we performed echocardiographic screen with transabdominal sonography. Blood flow was primarily seen in the right ventricle with little color signal noted in the left ventricle (Fig. 1). Two dimensional imaging suggested a left ventricle size which was smaller than that of the right ventricle (4.3 × 3.0 mm vs. 4.9 × 3.3 mm respectively) with a balloon-shaped contour to the left ventricle, hyperechogenicty of the left ventricular walls and poor left ventricular function (Fig. 2). In the three vessel trachea view (3-VT) with color Doppler, a narrowed reverse blood stream signal was observed in the aortic arch, and the narrowest blood stream signal was 0.7 mm in width. In the meantime, widened blood stream signal was observed in the pulmonary artery (2.2 mm) and the ductus arteriosus (1.8 mm) (Fig. 3). Hence, a diagnosis of HLHS with EFE was established. After thorough consultation, the parents decide to terminate pregnancy at 12 weeks' gestation. Autopsy revealed hypoplasia of ascending aorta and aortic arch. The internal diameter of the ascending aorta was 0.8 mm, while the internal diameter of

the narrowest part of the aortic arch, i.e. the aortic isthmus, was 0.5 mm. Meanwhile, the pulmonary artery and the ductus arteriosus was widened, with an internal diameter of 2.0 mm and 1.8 mm, respectively (Fig. 4A), and inspection of the aortic valve revealed aortic valve atresia (Fig. 4B). Besides, apparently narrowed mitral valve annulus and hypoplasia of leaflets was observed, which led to an almost complete mitral valve atresia (Fig. 4C). The endocardium of the left ventricle was thickened (Fig. 4D). Altogether, these finding confirmed the prenatal diagnosis. Chromosome analysis and microarray analysis revealed a 45X karyotype, thus established a diagnosis of Turner syndrome.

Discussion And Conclusions

Hypoplastic left heart syndrome (HLHS) was proposed by Lev in 1952 [5]. The etiology of this congenital condition remains largely unknown. However, it has been found that HLHS was closely related to various chromosome abnormalities, such as 45X, trisomy 13 syndrome and trisomy 18 syndrome. Meanwhile, several gene mutations have been related to HLHS, such as mutations of *NKX2-5*, *NOTCH1*, *ETS1*, *HAND1* and *rbFOX2* [6]. HLHS may be isolated or found in conjunction with Turner syndrome, Noonan Syndrome, Smith-Lemli-Optiz or Holt Oram.

Two types of HLHS can be observed in first trimester. Type I HLHS was characterized by atresia of mitral valve and aortic valve, and extremely hypoplasia of left ventricle. Meanwhile, type II HLHS was characterized by hypoplasia of mitral valve, severely stenosed or occluded to atretic, balloon-shaped left ventricle, and endomyocardial fibroelastosis of left ventricle. Most HLHS diagnosed in first trimester was type I HLHS. In 4-CV color Doppler scan may reveal a single blood stream in atrium and ventricle. In the 3-VT, a thin reverse blood stream may be observed in the aortic arch, the left ventricle may be difficult to be observed in 2D [3, 9]. Reports on Type II HLHS diagnosed in first trimester is extremely rare. For this type, findings in 4-CV and 3-VT color Doppler were similar with findings of type I HLHS. However, the left ventricle of type II HLHS was balloon-shaped, which would be readily recognized in 2D. besides to In addition, hyperechonic signals can be observed at the endocardium in fetus with type II HLHS.The autopsy revealed extreme mitral valve hypoplasia and aortic valve atresia, and rough endometrium of the left ventricle, all of which were consistent with sonographic findings at 11^{+2} weeks' gestation.This diagnosis of HLHS in the first trimester was readily achievable due to its characteristic presentation,. Most cases can be diagnosed employing the 4 -CV and 3-VT color Doppler. However, Change to However, some cases of HLHS may not be able to be diagnosed in the first trimester, as the cardiac abnormalities present require longitudinal growth of the fetus before the obvious asymmetry of the heart and vascular structures becomes visually apparent. Therefore some forms of HLHS cannot be completely excluded with a "normal" first rimester scan [10].

The morbidity of newborns with HLHS remains remains significant. The Norwood procedure has significantly increased the survival rate of patients with HLHS. However, the high cost of medical care, the need for multiple surgeries and the ongoing need for a lifetime of cardiac care and potential additional interventions may limit it from benefiting a larger population [11].Intrauterine treatment potentially

benefiting HLHS has had much focus, but the subset of HLHS fetuses which may potentially benefit from intervention remain limited and would not have been a potential modality to be utilized this case.

Declarations

Ethics approval and consent to participate

Not applicable

Consent for publication

Written informed consent (in simplified Chinese) for publication was obtained from the parents.

Availability of data and materials

Not applicable.

Conflict of interest

The authors declared that they have no conflicts of interest to this work.

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Authors' contributions

YSH and TXX designed the supervised this study. YZJ and PYL collected and refined the images. LMF, QGC and LXY drafted the manuscript. YSH revised and submitted this manuscript.

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Not applicable.

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Figures

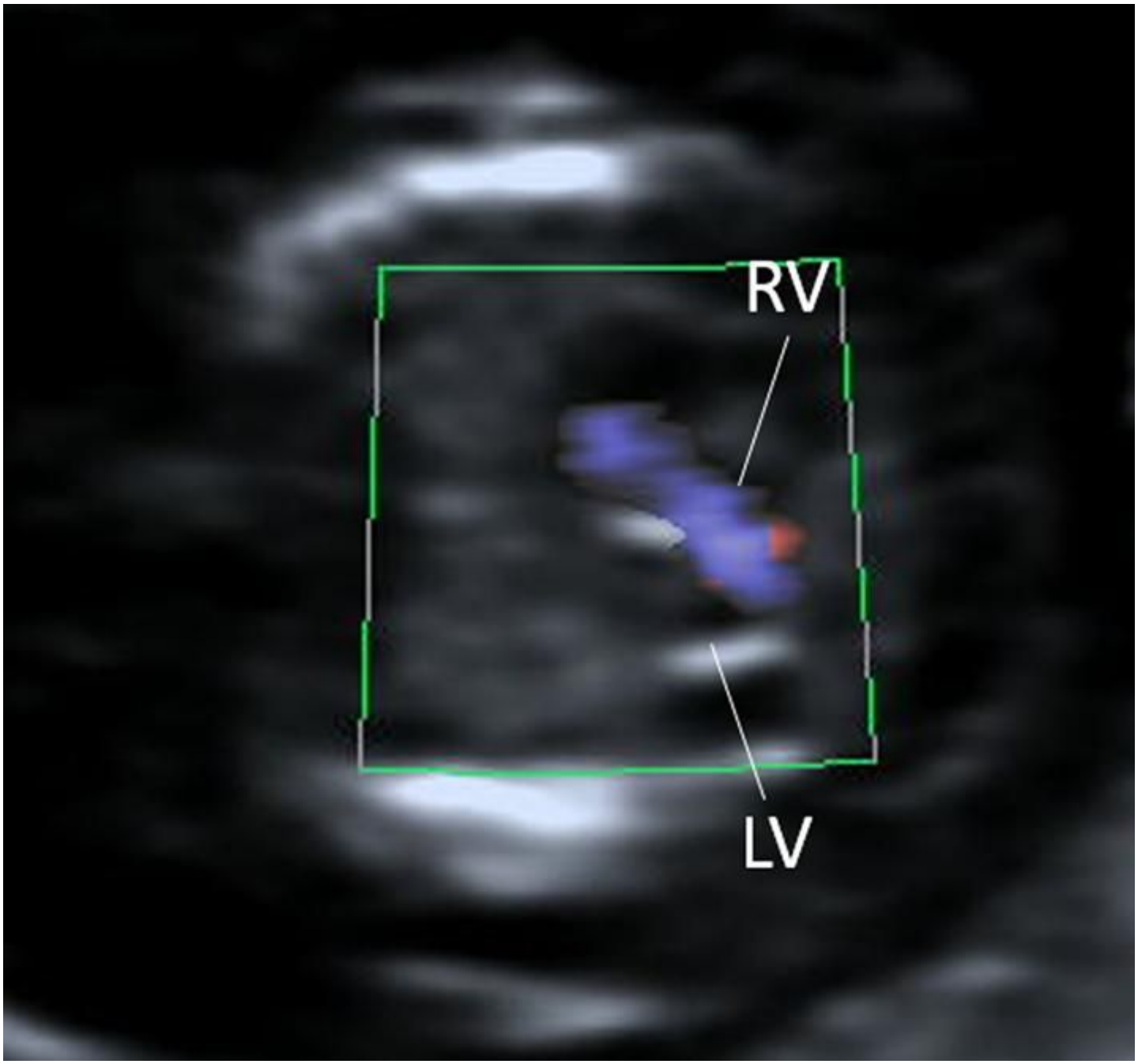

Figure 1

4-CV color Doppler Only one blood stream can be seen in the right side LV: left ventricle; RV: right ventricle

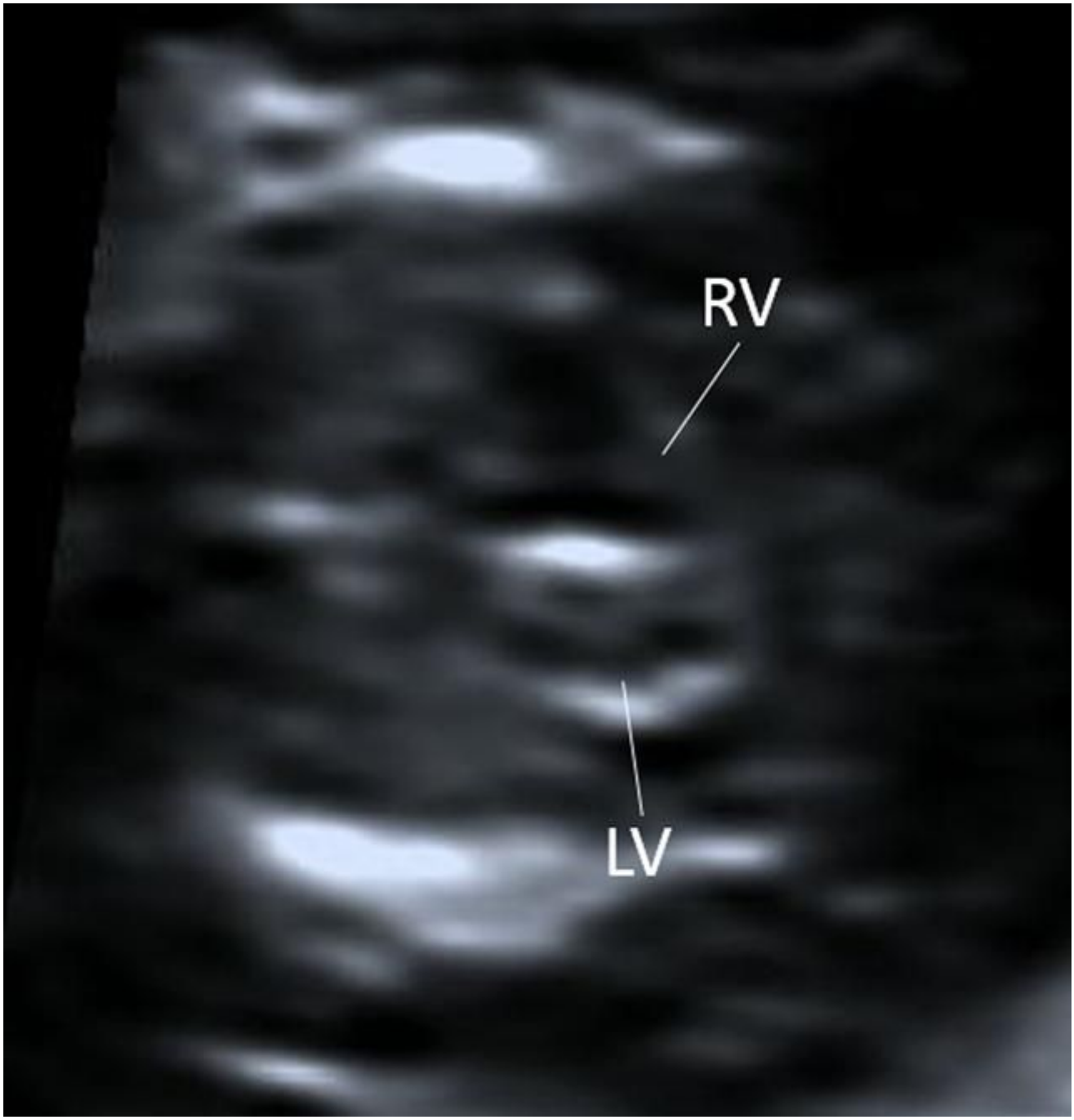

Figure 2

In 2D 4-CV Hyperechonic signals was observed at the endocardium of the left ventricle. ARCH: aortic arch; MPA: main pulmonary artery

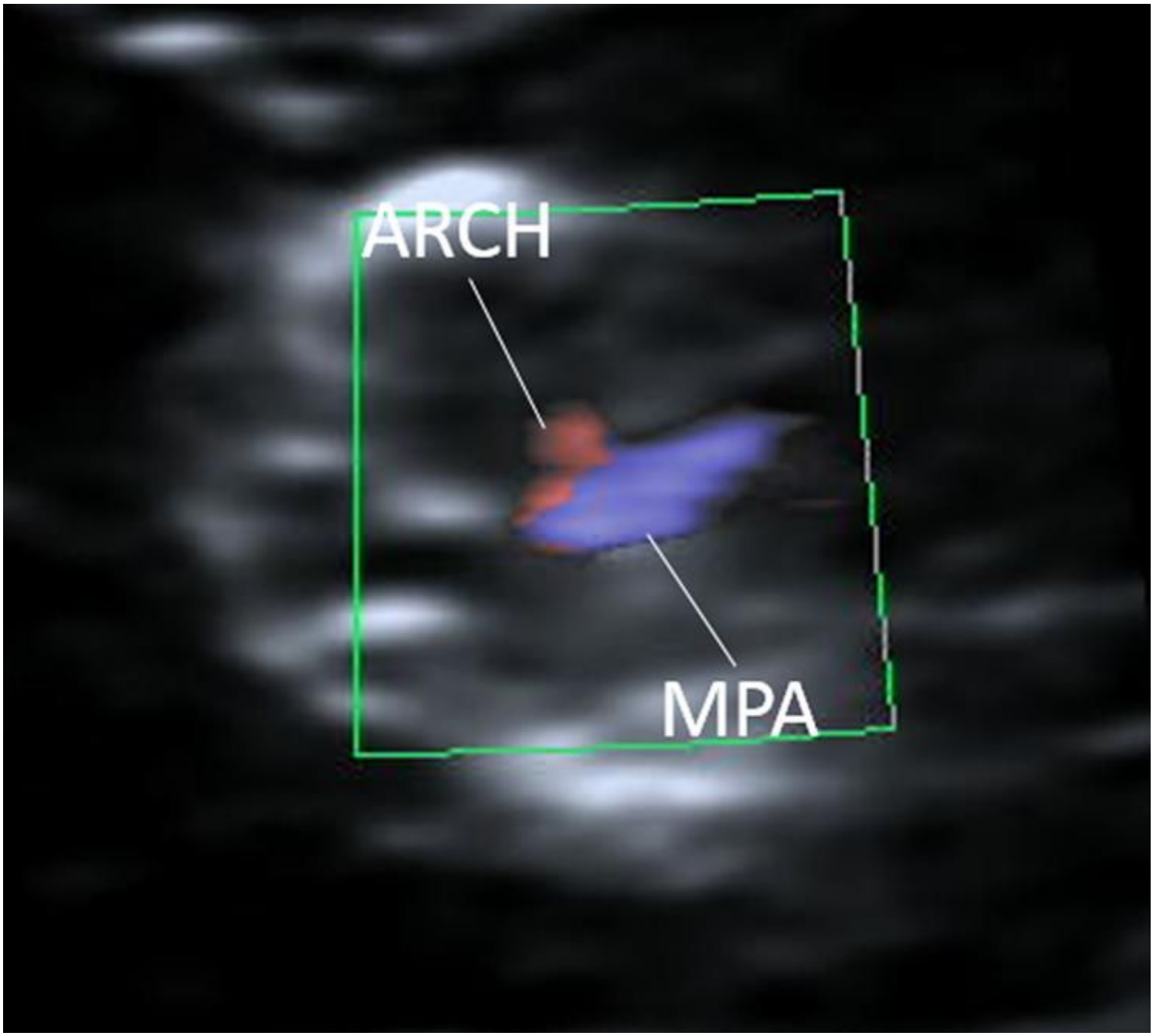

Figure 3

3-VT color Doppler A thin reverse blood stream was observed in the aortic arch. LV: left ventricle; RV: right ventricle

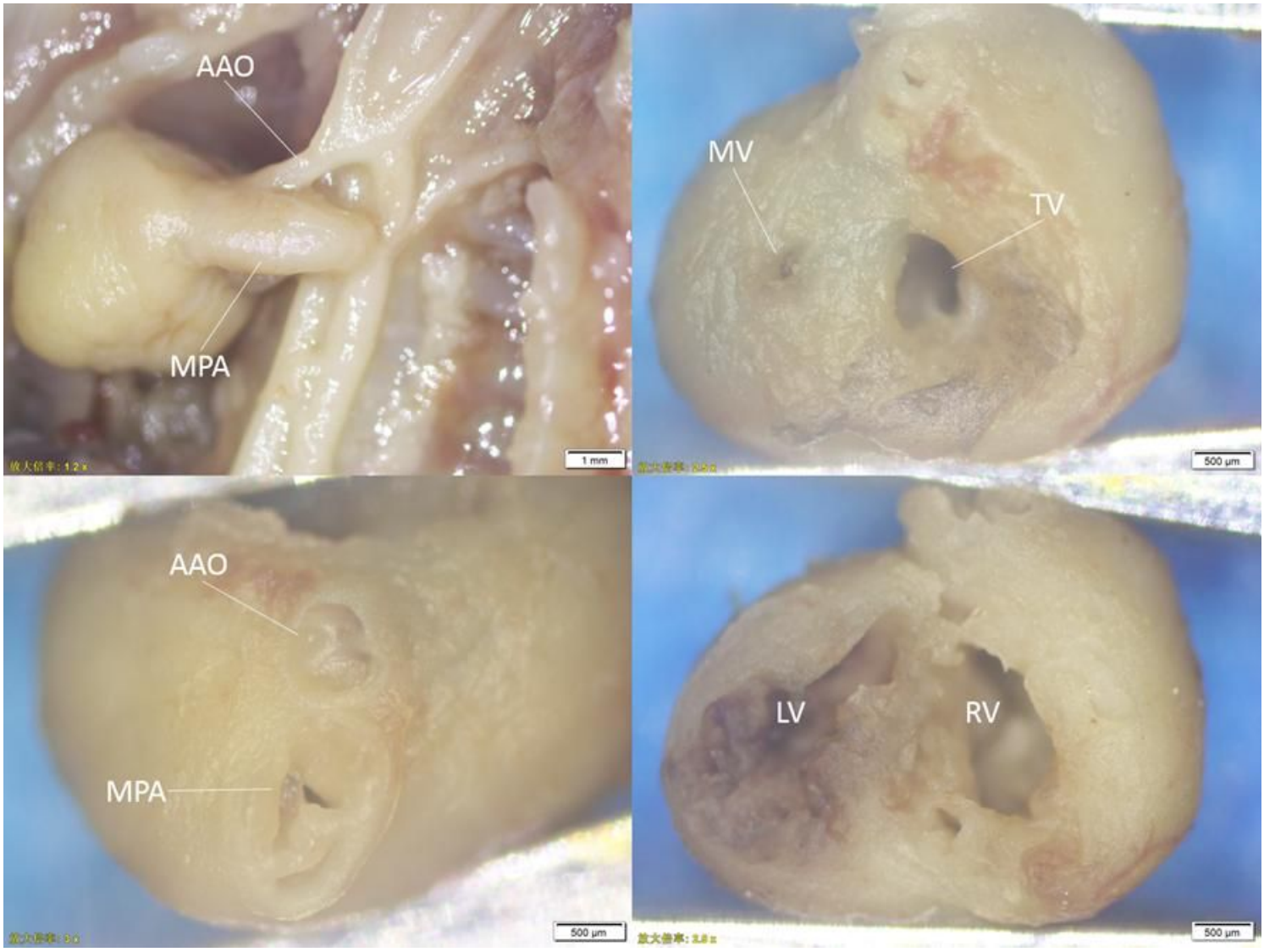

Figure 4

Autopsy of the heart from the aborted fetus A. The ascending aorta and arch of the aorta were hypoplastic, and the pulmonary and ductus arteriosus were compensatively widened (x 12). B. Mitral valve annulus was apparently stenosed, leading to a nearly occluded opening (x 25). C. Aortic valve was occluded (x 30). D. The endomyocardium of the left ventricle was rough in the transverse section of ventricle (x 25).