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CASE REPORT

Persistent truncus arteriosus with absent semilunar valve in first trimester

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Abstract
Persistent truncus arteriosus (PTA) is a relatively uncommon congenital heart disease, accounting for approximately 0.7–1.4% of all congenital cardiac abnormalities worldwide. PTA is usually accompanied by a single semilunar valve, with leaflets ranging from one to six in number. However, absent semilunar valve (ASV) is rarely seen in PTA. Here, we report a case of prenatally diagnosed PTA accompanied by ASV (PTA-ASV) confirmed by postmortem autopsy.

Keywords Persistent truncus arteriosus · Absent semilunar valve · Fetal echocardiography

Case summary

A 28-year-old pregnant woman, gravida 2, para 0, with no significant medical history and no family history of congenital heart defects was referred to our department for nuchal translucency (NT) scan at 13 + 3 weeks' gestation, which demonstrated increased fetal NT thickness (3.3 mm), ascites, and cardiac anomalies. The four-chamber view (4-CV) revealed cardiac enlargement, with a cardiothoracic ratio of 0.43. The outflow tract view revealed a single artery, which was continuous to the aortic arch, arising from the ventricles and overriding a large ventricular septal defect (VSD). Color Doppler of the outflow tract view revealed significant subarterial to-and-fro flow (Fig. 1). Genetic counseling was recommended to discuss the risk of aneuploidy, given the presence of a congenital heart defect. Based on these findings, the parents requested induction at 14 + 5 weeks' gestation, and consented to autopsy and karyotype analysis.

Fetal autopsy demonstrated absence of the thymus and significant structural cardiac anomalies. The common arterial trunk was enlarged and rode astride the VSD. Semilunar valve was absent, with a small residual valve incisura being observed. The extremely stenotic main pulmonary artery arose from the left posterior wall above the antrum of the common trunk. Ductus arteriosus was absent (Fig. 2). Hence, the definitive diagnosis in this case was type I PTA with ASV. Karyotype analysis revealed 22q11 deletion.

Discussion

Persistent truncus arteriosus (PTA), which is rarely encountered in clinical practice [1], is commonly accompanied by a single semilunar valve [2, 3]. However, absence of a semilunar valve (ASV) is relatively uncommon, which may manifest as absence of the pulmonary valve, aortic valve, or both [4–6]. Most studies on ASV found absence of the pulmonary valve, which would not lead to heart failure or fetal death and was more commonly diagnosed by ultrasound scanning in the second trimester [4]. Meanwhile, absence of the aortic valve and both valves were extremely rare, since most fetuses with these conditions would not survive in the first trimester due to severe heart failure [5, 6]. PTA is usually accompanied by an abnormal semilunar valve, with leaflets ranging from one to six in number [2, 3]. However, there is no report of PTA accompanied by ASV. Presumably, a fetus with this complex condition may have manifestations that are similar to those in a fetus with absence of the aortic valve, such as fetal edema and enlarged heart, which we also detected in this case. Diagnosis of ASV in the first trimester is based on the special signs on color Doppler and

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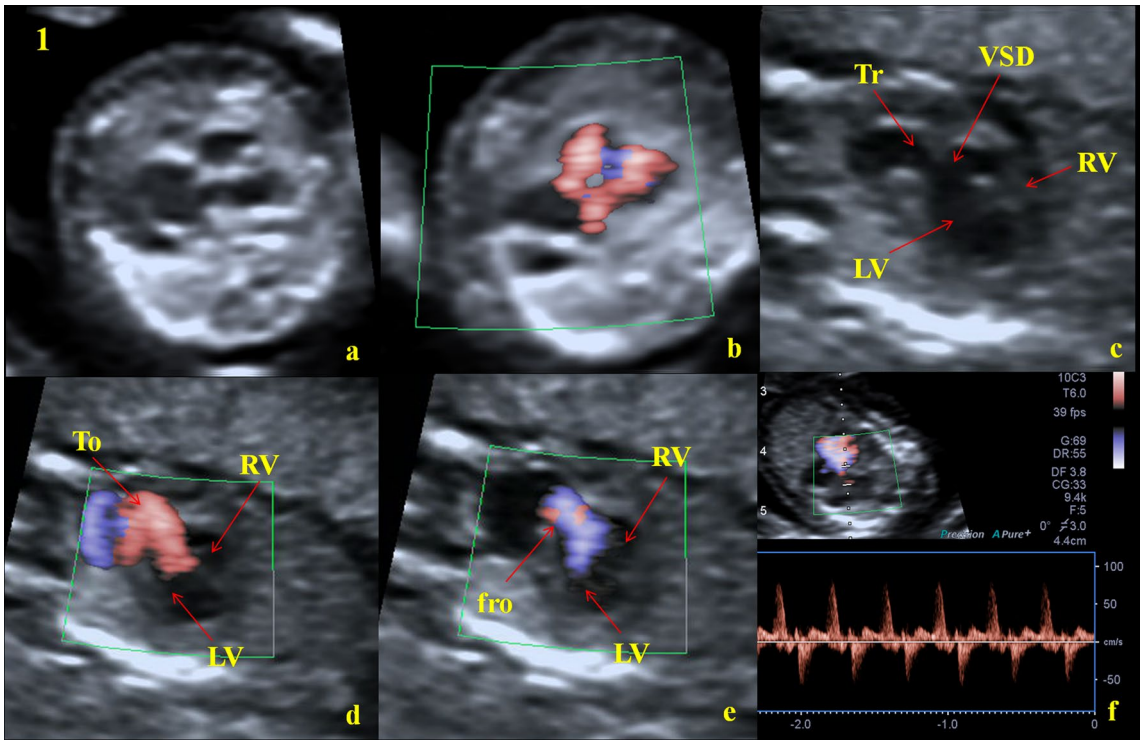

Fig. 1 Prenatal echocardiography of the fetus. **a** The 4-CV was structurally normal but enlarged. **b** Color doppler of the 4-CV was normal. **c** A trunk overrode the VSD, with distal large artery enlargement. **d**, **e** Color Doppler showed blood flow through the semilunar valve ring with to-and-fro sign. **f** Spectral doppler showed pansystolic turbulence and pandiastolic reflux spectrum. *Tr* trunk, *RV* right ventricle, *LV* left ventricle, *VSD* ventricular septum defect

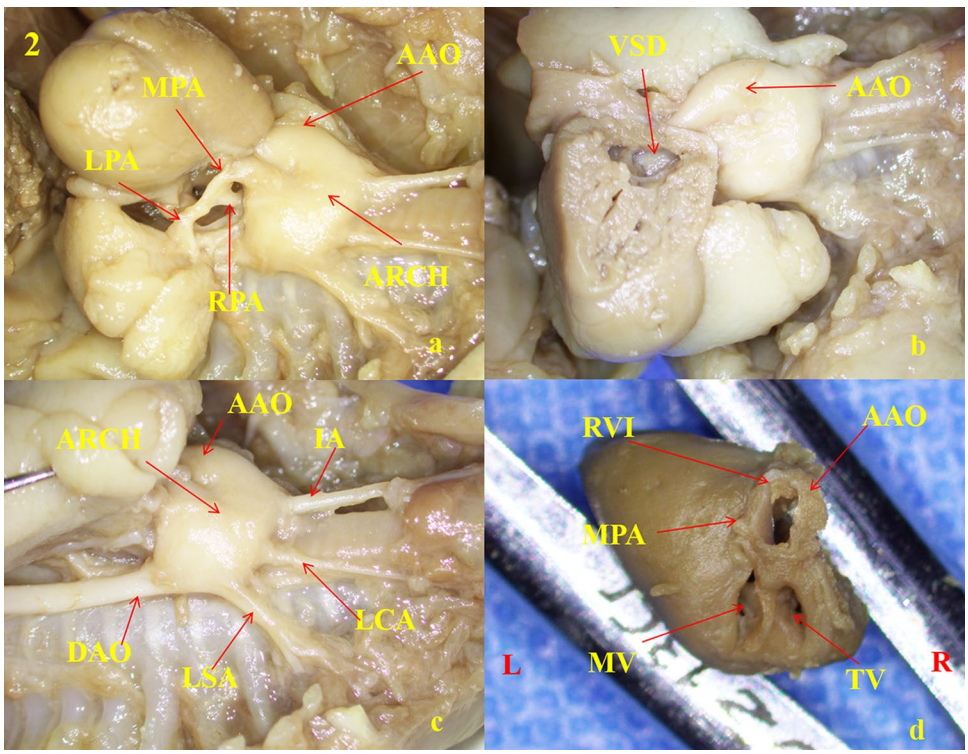

Fig. 2 Pathological finding of the fetus under stereomicroscope. **a** MPA originates from the left posterior wall above the antrum of the common trunk. **b** The trunk overrides the VSD. **c** The trunk was markedly enlarged before emptying into the aorta arch. **d** Absence of semilunar valve with a small residual valve incisura. *Tr* trunk, *AAO* ascending aorta, *ARCH* aortic arch, *DAO* descending aorta, *IA* innominate artery, *LCA* left coronary artery, *LSA* left subclavian artery, *MPA* major pulmonary artery, *LPA* left pulmonary artery, *RPA* right pulmonary artery, *TV* tricuspid valve, *MV* mitral valve, *VSD* ventricular septum defect, *RVI* residual valve incisura

spectral Doppler, i.e., "to-and-fro" signals detected in the corresponding artery. The special signs on color Doppler of the three-vessel and trachea views should be recognized in first trimester screening, especially in fetuses with heart failure [7].

In addition, autopsy revealed absence of the thymus, which is an extra cardiac anomaly typically associated with PTA and is often accompanied by chromosome 22q11 deletion [8]. Moreover, 22q11 deletion, which leads to various cardiovascular defects including PTA, was also found in karyotype analysis in this case [9]. PTA-ASV remains a challenge to clinicians since few reports to date have described its management and prognosis. For this rare condition, prenatal sonographic scanning of PTA may facilitate prognosis assessment and treatment planning.

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Compliance with ethical standards

Conflict of interest There are no financial or other relations that could lead to a conflict of interest.

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