

# Truncus Arteriosus With Absent Semilunar Valve: Prenatal Diagnosis and Morphology

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## Case report

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# Abstract

**Background** Truncus arteriosus (TA) is a rare cyanotic congenital heart defect that involves septation failure of the heart's main arterial outflow tract. TA is usually accompanied by a single semilunar valve. Varying morphologies of the truncal valve have been reported; however, an absent semilunar valve (ASV) is rarely seen in TA.

**Case presentation** We report the first fetus with trisomy 13 prenatally diagnosed with TA accompanied by ASV in the first trimester, and confirmed by anatomy.

**Conclusion** Fetal echocardiography is essential for prenatal diagnosis of TA accompanied by ASV.

## Introduction

We report on a 28-year-old primigravida, who was exposed to a newly-renovated working environment, and with no significant medical history and no family history of congenital heart disease was referred to our department for NT scan at 12<sup>+2</sup> weeks' gestation. Fetal echocardiography (Apolio 500; Canon Medical Systems Corporation, Shimoishigami, Otawara-Shi, Tochigi, Japan) was performed at 13<sup>+2</sup> weeks' gestation owing to fetal NT thickening (3.5 mm), which showed fetal edema, a wave inversion in the DV and cardiac anomalies.

## Case Presentation

The four-chamber view revealed right atrioventricular enlargement, and color Doppler revealed severe tricuspid regurgitation (Fig. 1a). The outflow tract view revealed a large VSD and a single arterial vessel without a definite semilunar valve (Fig. 1b). The three-vessel and tracheal views also showed a single artery with a stenotic vessel arising from the artery (Fig. 1c), and another vessel originating from the aortic arch, with an oblique course to the right shoulder (Fig. 1d). Color Doppler revealed significant "to-and-fro" flow (Fig. 1e and f) in the single arterial vessel. Spectral Doppler showed a "to-and-fro" spectrum in the single artery (Fig. 1g).

The parents opted for pregnancy termination after prenatal consultation at 14 weeks' gestation, and heart autopsy and whole-genome exon sequencing were performed after obtaining the parents' informed consent. At autopsy, the common arterial trunk was enlarged and over-rode the VSD. The leaflets of the semilunar valve were completely absent (Fig. 2a). The extremely stenotic main pulmonary artery arose from the posterior wall of the common trunk, and then bifurcated into left and right branches, and the fetal ductus arteriosus was absent (Fig. 2b). We also found that the fetus had an aberrant right subclavian artery (Fig. 2c) and left-hand ulnar polydactyly (Fig. 2d). Hence, the definitive cardiac diagnosis in this case was type I TA with ASV. Whole-genome sequencing demonstrated trisomy 13 (47, XY, +13).

## Discussion

TA is a rare cardiovascular malformation. The single truncal valve shows great morphological variability, with different numbers of leaflets and presenting frequently as dysplastic and insufficient or, more rarely, stenotic<sup>[1]</sup>. ASV is relatively uncommon, and may manifest as absence of the pulmonary valve, aortic valve, or both<sup>[2]</sup>. The pathogenesis of ASV is unclear, and best explained by underdevelopment of the endocardial cushion tissue at the ventriculoarterial junction<sup>[3]</sup>. In addition, genetic or environmental factors, hemodynamic changes, and mesenchymal cells of extracardiac origin derived from neural crest cells might play roles in the occurrence of ASV<sup>[4]</sup>. The presumed pathogenesis of TA with a dysplastic truncal valve is an insufficient volume of neural crest cells<sup>[4]</sup>. However, other theories considering the high frequency of combined malformations of the arterial valves and conotruncus suggest a common pathogenesis for these two conditions involving abnormalities of separation of the developing outflow tracts<sup>[5]</sup>. Recent studies indicated that valvulogenesis is a dynamic and multistep process likely involving many transcription factors and signaling pathways involving members of the TGF- $\beta$  superfamily, Notch, BMP and GATA families, NFATC1, Wnt/ $\beta$ -catenin, Twist-1, SOX9, and others<sup>[6]</sup>. These transcription factors and signaling pathways are essential for semilunar valve development.

We identified only one well-documented published case of TA accompanied by ASV that was also diagnosed in the first trimester<sup>[7]</sup>. We speculate that the reason for the rarity of this condition might be that fetuses with these conditions do not survive the first trimester owing to severe heart failure, similar to fetuses with an absent aortic valve or missing both semilunar valves<sup>[8]</sup>. In our case, the fetus had systemic edema, abnormal DV blood flow, tricuspid regurgitation, and cardiac enlargement, which might have been manifestations of early heart failure. We believe that with developments in ultrasonographic technology, more similar cases will be detected in the first trimester and the disease spectrum of congenital heart disease will be updated.

TA accompanied by other abnormalities is related to chromosomal abnormalities. The fetus in our case had trisomy 13, and it is the third most common type of aneuploidy after trisomy 21 and trisomy 18, and is associated with congenital heart defects and a poor prognosis<sup>[9]</sup>. Trisomy 13 may affect the structural integrity of the cardiac system, causing ventricular septal defects, atrial septal defects, patent ductus arteriosus, and other abnormalities. However, to our knowledge, we report the first fetus with trisomy 13 prenatally diagnosed with TA accompanied by ASV.

## Conclusion

TA accompanied by ASV remains a challenge to sonographers and clinicians because there is no reported treatment, and because the condition should be diagnosed in the first trimester. Echocardiographic findings of a single artery overriding a large VSD and the “to-and-fro” flow sign is essential in the diagnosis and helps when providing further pregnancy treatment and during consultation with the parents.

# Abbreviations

ARCH: aortic arch, ARSA: aberrant right subclavian artery, ASV: absent semilunar valve, CT: common trunk, DAO: descending aorta, DV: ductus venous, LA: left atrium, LCA: left carotid artery, LSA: left subclavian artery, LV: left ventricle, MPA: major pulmonary artery, NT: nuchal translucency, PA: pulmonary artery, RA: right atrium, RAA: right atrial appendage, RCA: right carotid artery, RV: right ventricle, TA: Truncus arteriosus, TR: tricuspid regurgitation, VSD: ventricular septal defect

# Declarations

## Declaration of competing interest

None.

## Ethics approval and consent to participate

The study protocol was approved by the ethics review board of Sichuan University. We have obtained written informed consent from the patient.

## Consent for publication

Not applicable.

## Availability of data and materials

The datasets used and/or analysed during the current study are available from the corresponding author on reasonable request.

## Competing interests

None.

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## Authors' Contributions

Lihong Pu and Xiaohui Dai contributed equally to this work.

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## Figures

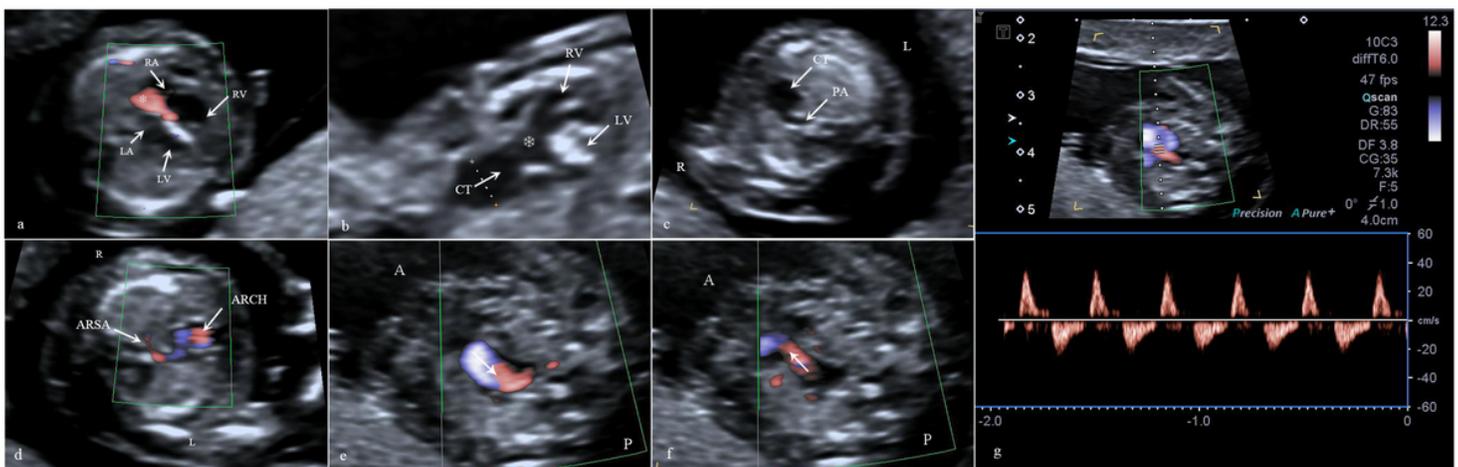
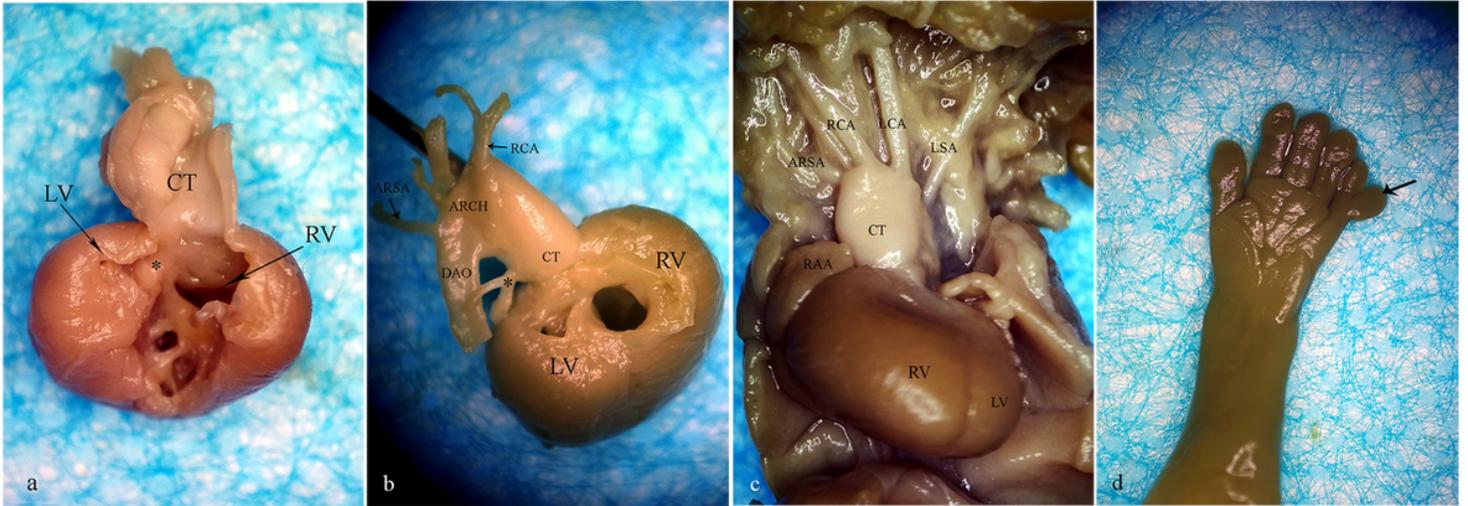


Figure 1

Prenatal fetal echocardiography. a Four-chamber view showing an enlarged right heart, and severe TR (asterisk). b Outflow tract view showing a large VSD (asterisk) and a common trunk. c Three-vessel and tracheal views showing a common trunk and PA. d The ARSA originates from the aortic arch, with an oblique course to the right shoulder. e The blue color in the Doppler images indicates antegrade flow in the common trunk during systole. f The red color in the Doppler images indicates retrograde flow in the common trunk during diastole. g Spectral Doppler showing the “to-and-fro” spectrum.



**Figure 2**

Gross pathology of the fetus under stereomicroscopy. a The enlarged trunk overrides the VSD (asterisk), and the leaflets of the arterial valve are completely absent. b The right ventricle is enlarged, and the MPA (asterisk) originates from the posterior wall of the common trunk. c The ARSA originates as a separate fourth branch of the aortic arch. d Ulnar polydactyly of the left hand (arrow).