Cardiovascular Ultrasound

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# Prenatal diagnosis of pulmonary atresia with intact ventricular septum: a single-center study in China

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## Abstract

**Objectives** To evaluate the efficacy of prenatal ultrasound in diagnosing pulmonary atresia with intact ventricular septum (PA/IVS).

**Methods** This retrospective study analyzed 48 cases of PA/IVS at the Fujian Maternity and Child Health Hospital between January 2013 and December 2023. Prenatal ultrasound was used to characterize and classify the features of PA/IVS. Pregnancy outcomes were followed up, and the results were compared with post-termination pathological anatomical findings or postnatal imaging. This study aims to enhance the understanding of PA/IVS and improve the accuracy of its prenatal diagnosis.

**Results** Among the 48 PA/IVS cases, four were diagnosed during early pregnancy and 44 during mid-to-late pregnancy. In the mid-to-late pregnancy group, there were 29 cases of type I (TV-Z scores ranging from − 1.77 to 5.22), 10 cases of type II (TV-Z scores ranging from − 3.50 to -2.06), and five cases of type III (TV-Z scores ranging from − 4.29 to -7.41). The cohort included 41 singleton pregnancies and seven twin pregnancies. Ventriculo-coronary artery communication (VCAC) was observed in 19 cases. Additional abnormalities included Ebstein's anomaly (EA) in three cases, restricted opening of the foramen ovale in one case, increased inner diameter of the foramen ovale in one case, reversal or deepening of the a-wave of the ductus venosus in six cases, and umbilical vein pulsation in one case. Genetic testing (amniocentesis, NIPT, or SNP-array) was performed in 19 cases, with one case revealing a genomic copy number deletion in the q22.3 region of chromosome 21. Pregnancy outcomes included 41 terminations (five with pathologic dissection or vascular casting), five live births, one selective reduction, and one intrauterine death.

**Conclusion** Fetal echocardiography is an effective tool for diagnosing PA/IVS. While PA/IVS can be diagnosed in early gestation, it remains diagnostical challenging. Given the progressive nature of PA/IVS in utero, sequential ultrasound examinations during the second and third trimesters are essential for monitoring disease progression and hemodynamic changes. Additionally, a comprehensive evaluation for associated intracardiac and extracardiac anomalies should be systematically conducted throughout the pregnancy.

**Keywords** Fetus, Prenatal ultrasound, Pulmonary atresia with intact ventricular septum, Pregnancy outcome

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**Graphical Abstract**


Prenatal ultrasound

early pregnancy (n=4)

mid-to-late pregnancy (n=44)
- 29 cases of type I
- 10 cases of type II
- 5cases of type III

Associated malformations

VCAC (n=19)
EA (n=3)
21q22.3 microdeletion (n=1)

Pregnancy outcomes

terminations (n=41)
live births (n=5)
selective reduction (n=1)
intrauterine death (n=1)

PA/IVS is detectable prenatally but requires sequential scans to monitor progression

## Introduction

Pulmonary atresia with intact ventricular septum (PA/IVS) is a rare cyanotic congenital heart defect by the complete closure of the pathway from the right ventricle to the pulmonary artery, while the interventricular septum remains intact. It accounts for approximately 1–3% of all congenital heart diseases [1]. The pathological anatomy of PA/IVS is highly variable, involving malformations of the pulmonary valve, right ventricle, tricuspid valve, and coronary arteries. Additionally, pulmonary atresia may develop progressively with advancing gestational age, as severe pulmonary stenosis can get involved into pulmonary atresia during fetal development [2]. The heterogeneity of its pathological anatomy and the potential for intrauterine progression present significant challenges to the accuracy of prenatal diagnosis.

The long-term prognosis of PA/IVS depends primarily on the anatomical features and the surgical repair approach. Numerous studies have attempted to identify predictive factors during the fetal period that can determine the postnatal circulation type [3–5]. With advancements in medical science and technology, fetal cardiac intervention has emerged as a specialized field. In-utero pulmonary valvuloplasty can be performed in fetuses with PA/IVS to relieve obstructive lesions, promote right ventricular development, and increase the likelihood of achieving biventricular circulation after birth [6].

PA/IVS is a congenital heart defect characterized by ductus arteriosus (DA)-dependent pulmonary circulation. After birth, pulmonary blood flow relies on to the patency of DA. Closure of the DA leads to severe hypoxemia, necessitating prostaglandin E1 infusion in all newborns with PA/IVS to maintain DA patency [7]. Accurate prenatal ultrasound diagnosis and risk stratification are therefore critical for effective prenatal counseling, perinatal management, and postnatal treatment planning [8]. Although numerous studies have reported on the prenatal ultrasound characteristics of PA/IVS in mid-to-late pregnancy, the condition can still be missed or misdiagnosed by less experienced clinicians. Early diagnosis during pregnancy is crucial, as it provides ample time for clinical counseling and informed decision-making, thereby reducing physical and psychological trauma for pregnant women.

Therefore, it remains crucial to investigate how prenatal ultrasound can accurately diagnose PA/IVS throughout pregnancy and enhance the integrated prenatal-to-postnatal management of affected fetuses. In this retrospective study, we included 48 fetuses diagnosed with PA/IVS via prenatal ultrasound at various stages of pregnancy, from early to late gestation. We analyzed

the prenatal ultrasound features of PA/IVS, classified the cases into subtypes, and tracked their pregnancy outcomes. These outcomes were compared with post-labor induction pathological findings or postnatal imaging results to deepen the understanding of PA/IVS and improve the accuracy of prenatal ultrasound diagnosis.

## Materials and methods

### Study population

From January 2013 to December 2023, a total of 72 cases were diagnosed with PA/IVS and enrolled in this study. Prenatal ultrasound findings were compared with post-termination pathological results or postnatal diagnostic findings. Due to incomplete ultrasound data or loss to follow-up, only 48 patients were included in the final analysis. Inclusion criteria required a prenatal ultrasound diagnosis of PA/ IVS plus either (1) pathological confirmation after labor induction, (2) postnatal diagnosis via Computed tomography angiography (CTA) or echocardiogram, or (3) complete ultrasound measurements for definitive diagnosis. All sonographers were certified prenatal specialists, and each PA/IVS diagnosis was confirmed by at least two physicians (including an associated chief physician or higher) via double-blind assessment. The study was approved by the Ethics Committee of Fujian Maternity and Child Health Hospital (2024KY309), with informed consent obtained from the families; in termination cases, separate consent was provided for pathological autopsy or autopsy cast verification.

### Fetal echocardiography

PHILIPS IU22 and GE Voluson S8, E8, and E10 ultrasound systems were used, with a probe frequency of 4.0 ~ 8.0 MHz. Obstetric and fetal cardiac examinations were performed to assess fetal growth and systemic development. In early pregnancy ($11–13^{+6}$weeks), the four-sectional method - including the upper abdominal transverse section, four-chamber (4 C) view, three vessel–trachea (3VT) view, and bilateral subclavian artery view - was employed for fetal cardiac assessment [9, 10]. During mid to late pregnancy, fetal heart scanning followed segmental sequential analysis per the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) guidelines [11]. The 4 C and right ventricular outflow tract (RVOT) views were used to assess right ventricular inflow, trabeculae, and funnel development; the RVOT and 3VT views evaluate right ventricular outflow tract patency, pulmonary valve motion, and main pulmonary artery/branch development. Additionally, color Doppler 4 C and ventricular short-axis views were analyzed for tricuspid regurgitation (TR) and ventriculocoronary arterial communications (VCAC).

PA/IVS in mid-to-late pregnancy was classified into three types based on right ventricular development and Z-score of the tricuspid valve (TV-Z) [12, 13]. Type I featured normal/mildly hypoplastic right ventricle with preserved inflow, trabecular, and outflow portions (TV-Z ≥-2); Type II showed moderate hypoplasia with myocardial hypertrophy obliterating leaving only inflow and outflow portions (TV-Z -4 to -2); Type III demonstrated severe hypoplasia with only the inflow portion remaining (TV-Z ≤-4).

### Integrated prenatal-postnatal management of PA/IVS fetuses

For fetuses diagnosed with PA/IVS, we conducted multidisciplinary consultations and recommended genetic testing, including non-invasive prenatal testing (NIPT) for chromosomal aneuploidy screening, followed by amniocentesis or percutaneous umbilical cord blood sampling for G-banded chromosome karyotyping and single nucleotide polymorphism (SNP) microarray after genetic counseling and informed consent. In cases of pregnancy termination, PA/IVS fetuses were examined using combining in situ observation and ex vivo immobilization [14], with cardiovascular casting performed via thoracic and abdominal cavity casting. For continuing pregnancy, we established a dedicated diagnostic and treatment pathway, including regular ultrasound monitoring, refined prenatal risk stratification, and comprehensive counseling on prognosis and perinatal management. Postnatally, echocardiography was performed using PHILIPS EPIQ 7 C and IE Elite systems (3.0 ~ 5.0 MHz probe frequency), following the American Society of Echocardiography (ASE) pediatric guidelines for segmental heart analysis [15], while infants with prenatal PA/IVS diagnosis underwent additional Computed tomography angiography CTA using a GE revolution CT (256 slices).

### Statistical analysis

The data were analyzed using SPSS 25.0 (IBM Corp). Prenatal ultrasound findings were systematically compared with postnatal outcomes, including imaging results, surgical findings, pathological anatomy, and cardiovascular casting results. Continuous variables are presented as mean ± standard deviation ($\bar{x} \pm s$), while categorical data are expressed as frequencies and percentages.

## Results

A total of 48 PA/IVS cases were included (Fig. 1), with the maternal age ranging from 21 to 41 years and gestational age ranging from 12.6 to 33.9 weeks. Most cases (44/48, 91.7%) were diagnosed in mid-to-late pregnancy, while only 4 (8.3%) were detected early. The cohort included 41 singleton pregnancies and 7 twin pregnancies (5 with

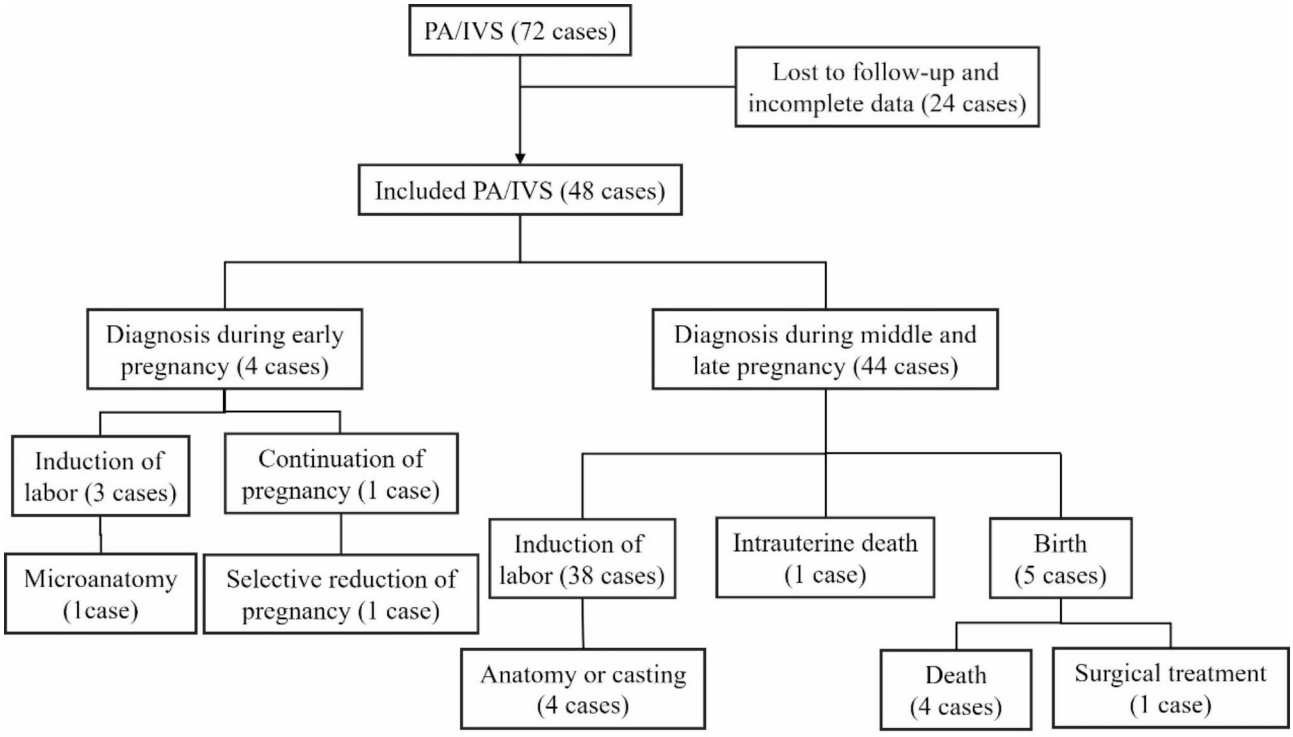


**Fig. 1** Flowchart of the cohort study

**Table 1** The condition of PA/IVS fetuses in early pregnancy

| Case | Gestational week | Cardiac findings | Diagnosis | Extracardiac abnormalities | Outcome |
|---|---|---|---|---|---|
| 1 | 13w0d | Hypoplastic RV. Retrograde ductus arteriosus flow. Multiple bidirectional turbulent flows within hypertrophied RV myocardium. Pericardial effusion (0.19 cm). | PA/IVS, VCAC, PE | NT thickening(3.2 mm), SUA | Termination, microanatomy |
| 2 | 13w0d | Enhanced echogenicity of the tricuspid valve with slightly restricted opening. Moderate TR. Retrograde ductus arteriosus flow with smaller diameter than aortic flow. | PA/IVS | Reversed DV a-wave | PA/IVS with VCAC confirmed by echocardiography at 18 weeks. Selective fetal reduction. |
| 3 | 12w4d | Hypoplastic RV. Retrograde ductus arteriosus flow with smaller diameter than aortic flow. Multiple bidirectional turbulent flows within hypertrophied RV myocardium. | PA/IVS, VCAC | Reversed DV a-wave | Termination |
| 4 | 13w5d | Dilated right atrium and ventricle. Severe TR. Retrograde ductus arteriosus flow. Pericardial effusion (0.2 cm). | EA, PA/IVS, PE | NT thickening (7.0 mm), generalized cutaneous edema, nasal bone dysplasia | Termination |

RV: right ventricle; PA: pulmonary artery; PE: pericardial effusion; TR: tricuspid regurgitation

one affected fetus and 1 with both fetuses affected). Pregnancy outcomes were as follows: 41 terminations (including 5 with pathological dissection/vascular casting), 1 selective reduction in twins, 1 intrauterine fetal death, and 5 live births. Among the live births, 4 resulted in neonatal death and 1 underwent percutaneous balloon pulmonary valvuloplasty.

#### Early pregnancy PA/IVS cohort ($n=4$)

The four early-diagnosed PA/IVS cases (mean gestational age: 13.07 ± 0.47 weeks; range: 12w4d-13w5d) showed mean crown rump length (CRL) of 6.83 ± 0.66 cm and mean nuchal translucency (NT) of 3.33 ± 2.6 mm (Table 1). Extracardiac anomalies included NT thickening (50%), reversed ductus venosus (DV) a-wave (50%), nasal bone hypoplasia (25%), skin edema (25%), ascites (25%), and single umbilical artery (SUA) (25%).

Intracardiac findings revealed Ebstein's anomaly (EA) (25%), VCAC (50%), and pericardial effusion (50%). All cases demonstrated: (1) absent pulmonary artery forward flow with ductus arteriosus (DA)-derived retrograde flow in 3VT view (pulmonary artery diameter < aorta; Fig. 2); (2) 4 C view findings - right heart enlargement ($n = 1$), hypoplastic right ventricle/thickened wall with left ventricular apex dominance ($n = 3$).

#### Mid-to-late pregnancy PA/IVS cohort ($n = 44$)

The 44 cases diagnosed between 19.0 and 33.9 weeks gestation demonstrated characteristic findings: thickened, hyperechoic pulmonary valves with absent leaflet motion and no antegrade flow on color Doppler, with ductus arteriosus (DA) derived retrograde perfusion (Fig. 3). Right ventricular morphology varied: 38 cases showed RV hypoplasia with wall thickening/hypokinesis, while 6 maintained normal RV dimensions. Tricuspid regurgitation was moderate-severe in 36 cases (mean velocity 364.5 ± 78.2 cm/s; range 134.1–500.3 cm/s), with mild/absent TR in 8 cases. Thirty-nine cases (88.6%) were membranous atresia presenting as well-developed pulmonary valve annulus and the main trunk of the pulmonary artery, the right ventricular outflow tract could be clearly identified; five cases (11.4%) were muscular atresia presenting as hypertrophied right ventricular outflow tract, the pulmonary valve was not clearly recognized, and the pulmonary valve annulus and pulmonary trunk were small. The 44 PA/IVS cases included 29 type I, 10 type II, and 5 type III cases. (1)29 cases of type I: right ventricular inflow, trabecular and funnel portions were present, with the TV-Z ranging from – 1.77 to 5.22, of which VCAC was present in 4 cases (4/29, 13.8%). (2)10 cases of type II: right ventricular inflow and funnel portion were present, trabecular portion was absent, with the TV-Z ranging from – 3.50 to -2.06, of which VCAC was present in eight cases (8/10, 80.0%). (3)5 cases of type III: only small inflow in the right ventricle, absence of trabecular and funnel portions, with the TV-Z ranging from – 4.29 to -7.41, of which VCAC was present in four cases (4/5, 80.0%).

Of the 44 PA/IVS cases, 12 were first examined at our hospital, while 24 referred from external consultations or outside hospitals had ultrasound diagnoses inconsistent with ours, including misclassified cases of pulmonary atresia with ventricular septal defect, critical pulmonary stenosis (CPS), and tricuspid valve dysplasia. The remaining six cases underwent early pregnancy screening at our hospital: one case showed NT thickening, cystic hygroma, generalized skin edema, and moderate

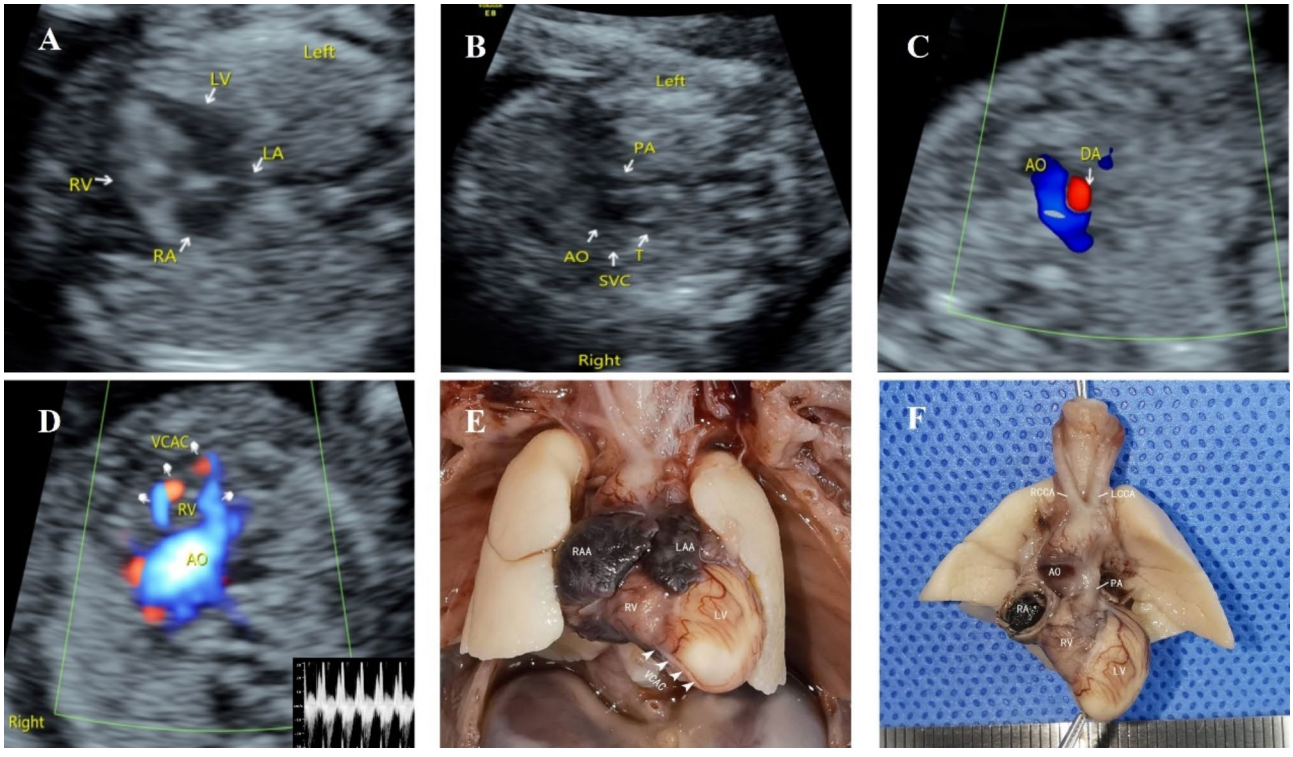


**Fig. 2** Prenatal ultrasound and postmortem findings in a 13-week PA/IVS fetus: (**A**) Four-chamber view demonstrating hypoplastic right ventricle with thickened wall; (**B**) Three-vessel-trachea (3VT) view showing pulmonary artery (PA) significantly smaller than aorta; (**C**) Retrograde ductus arteriosus (DA) flow with smaller diameter than aortic flow; (**D**) Multiple bidirectional turbulent flows within hypertrophied RV myocardium; (**E**) Specimen frontal view revealing hypoplastic RV (apical portion formed by left ventricle) with dilated epicardial coronary arteries; (**F**) Anatomical confirmation of marked hypoplastic PA compared to aorta

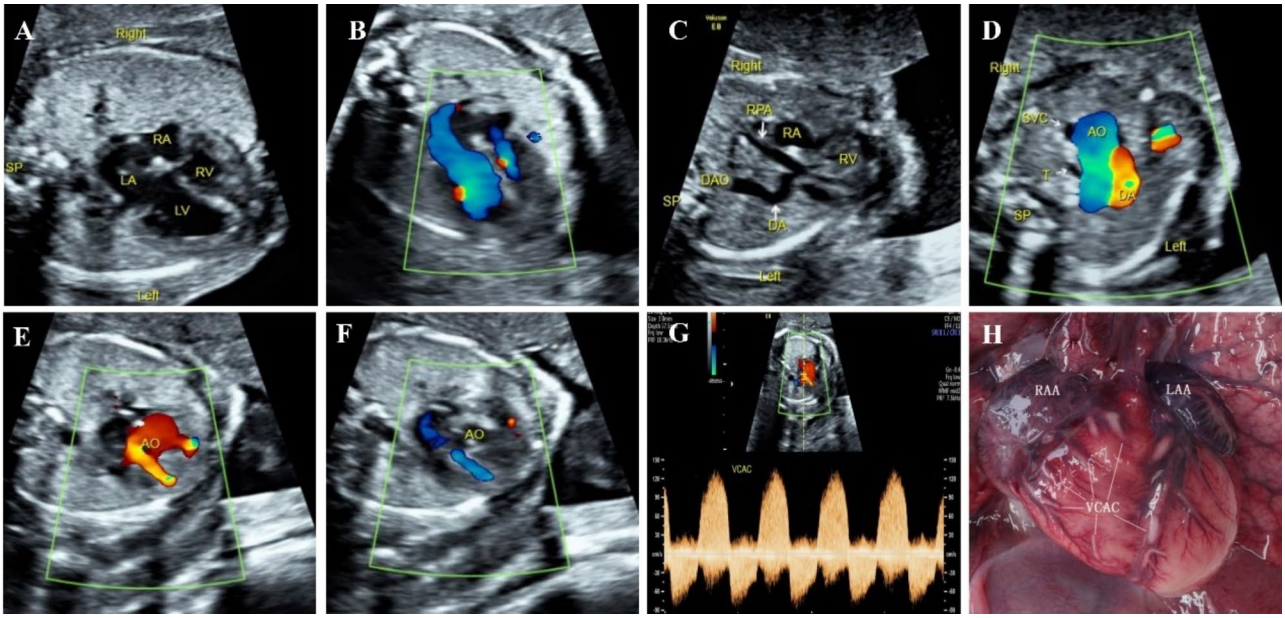


**Fig. 3** Imaging and pathologic features of a PA/IVS fetus ($23^{+3}$ weeks). (**A**) Four-chamber view showing hypoplastic right ventricle, dilated left atrium and left ventricle, and thickened right ventricular wall; (**B**) Absence of significant tricuspid regurgitation; (**C**) Small pulmonary valve annulus with tortuous ductus arteriosus (DA); (**D**) Retrograde DA flow into PA; (**E-G**). Color and spectral Doppler demonstrating VCAC. (**H**). Anterior specimen view revealing multiple RV surface VCACs

tricuspid regurgitation; one had a reversed a-wave in the DV; one presented with NT thickening, mild tricuspid regurgitation, and a reversed a-wave in DV; two displayed a pulmonary artery flow bundle smaller than the aorta's; one case had no obvious abnormalities. And two fetuses from a monochorionic diamniotic (MCDA) twin pregnancy progressed from initial pulmonary stenosis (PS) to pulmonary atresia with advancing gestational age.

#### Twin pregnancies in PA/IVS

Of the 48 PA/IVS fetuses, 41 (85.4%) were detected as singleton pregnancies and 7 (14.6%) were detected as twin pregnancies (Table 2). Case 4 was diagnosed with stage II twin-twin transfusion syndrome (TTTS) at 17 weeks of gestation, progressing to stage III TTTS one week later, and one fetus progressed from PS to PA/IVS, which then underwent ultrasound-guided fetoscopic vascular coagulation of placental surface traffic vessels. Postoperatively, the PA/IVS fetus developed VCAC, resulting in intrauterine fetal death one week later. As the gestational age increased, the other fetus developed ineffective intracardiac circulation, progressing from PS to pulmonary atresia, and PA/IVS was confirmed by postnatal echocardiography.

#### Ventriculo-coronary arterial communication in PA/IVS

VCAC was present in 19 of 48 PA/IVS fetuses (19/48, 39.6%) (Table 3). Ultrasound demonstrated multiple bidirectional turbulent flow signals between the aortic root and the hypertrophied right ventricular wall, with spectral Doppler revealed a continuous turbulent spectrum throughout the cardiac cycle. A case of PA/IVS diagnosed in early pregnancy developed VCAC at 18 weeks in a dichorionic diamniotic (DCDA) twin pregnancy, leading to selective reduction. Another case of PA/IVS at 23 weeks, presented with VCAC at 33 weeks and resulted in termination of pregnancy.

#### Combined malformations of PA/IVS

Among 48 PA/IVS fetuses, three (6.3%) had Ebstein's anomaly (EA). Other associated abnormalities included restricted foramen ovale opening (2.1%), enlarged foramen ovale diameter (2.1%), reversed/abnormal DV a-wave (12.5%), and umbilical vein pulsation (2.1%).

#### Genetic findings of pulmonary atresia with intact ventricular septum

Of 48 cases, genetic testing was performed in 19 cases. 10 underwent amniocentesis, 5 underwent non-invasive prenatal testing (NIPT), 15 received single nucleotide polymorphism (SNP) array analysis, 29 had no genetic testing. Karyotype results were normal. In one case (1/15, 6.7%), a 21q22.3 microdeletion was identified by microarray analysis, with no other pathogenic variants detected.

#### Prognosis of PA/IVS

Among four PA/IVS cases diagnosed at $11–13^{+6}$ weeks, three (75%) underwent early pregnancy termination. Pathologic examination of one case revealed micropathological findings: a hypoplastic right ventricle with left

**Table 2** Twin pregnancies in PA/IVS fetuses

| Case | chorionic and amniotic | Fetus with PA/IVS(Fetus A) | Fetus B | Characteristics of the case | Outcome |
|---|---|---|---|---|---|
| 1 | MCDA | PA/IVS, severe tricuspid regurgitation | (-) | / | Born at $34^{+4}$ weeks, Fetus A: 2340 g, Apgar 9-10-10, postnatal echocardiography: PA/IVS and restricted tricuspid valve opening with severe regurgitation. Fetus B: 2010 g, Apgar 7-10-10, postnatal echocardiography: patent ductus arteriosus and patent foramen ovale. |
| 2 | MCDA | PA/IVS, severe tricuspid regurgitation | VSD, ARSA | Reversed DV a-wave of two fetuses during early pregnancy | Termination |
| 3 | MCDA | PA/IVS, severe tricuspid regurgitation, increased cardiothoracic ratio, Reversed DV a-wave, ascites, excessive amniotic fluid | (-) | / | Born at $31^{+6}$ weeks, Fetus A: 1470 g, Apgar 10, postnatal echocardiography: PA/IVS, patent ductus arteriosus (left-to-right shunt), patent foramen ovale, Atrial septal defect, mitral regurgitation +-++, tricuspid regurgitation ++++, and the estimated systolic pressure of the pulmonary artery was 53mmHg; Fetus B, 1410 g, Apgar 10. |
| 4 | MCDA | Increased cardiothoracic ratio, severe tricuspid regurgitation, PA/IVS, Reversed DV a-wave, abnormal pulsation of the umbilical vein | Progression to PA/IVS, severe tricuspid regurgitation with progression of gestational weeks | TTTS, PA/IVS with increasing gestational weeks | Fetus A died in utero. Fetus B was born at 27w, 915 g, Apgar 10, Postnatal Echocardiography: PA/IVS, severe tricuspid regurgitation, patent ductus arteriosus (left-to-right shunt), patent foramen ovale, and the estimated systolic pressure of the pulmonary artery was 32mmHg |
| 5 | MCMA | PA/IVS, severe tricuspid regurgitation | (-) | / | Termination |
| 6 | DCDA | PA/IVS, right ventricular dysplasia, VCAC | (-) | Detected at 13 weeks of gestation | Selective reduction of fetus A, full-term delivery of fetus B |

ventricular apex dominance, dilated epicardial coronary arteries, and significantly narrowed pulmonary arteries relative to the aorta (Fig. 2). The remaining case, a DCDA twin pregnancy, was confirmed by echocardiography at 18 weeks and underwent selective reduction. Of 44 PA/IVS mid-to-late pregnancy diagnoses, 38 (86.4%) resulted in termination, including four with autopsy-confirmed pulmonary atresia (failed right ventricular outflow tract probe insertion). Postnatal echocardiography confirmed PA/IVS in five cases (11.3%) (Fig. 4): two with CTA-documented right coronary artery atresia died without intervention, two died after treatment withdrawal, and one survived following successful balloon pulmonary valvuloplasty. One case (2.3%) resulted in intrauterine fetal demise.

## Discussion

PA/IVS represents a complex spectrum of congenital heart disease with significant anatomic and functional heterogeneity. Our findings demonstrate that meticulous prenatal echocardiographic evaluation can reliably characterize the key features of this condition, including the critical distinction between membranous (88.6%) and muscular (11.4%) atresia subtypes. This differentiation carries important therapeutic implications, as only membranous atresia may be amenable to fetal pulmonary valvuloplasty [7, 16]. The progressive nature of right ventricular outflow tract obstruction (RVOTO) in some cases underscores the necessity for serial monitoring, particularly in twin pregnancies where hemodynamic disturbances may accelerate disease progression.

The early pregnancy detection rate of 8.3% in our cohort aligns with emerging evidence supporting first-trimester cardiac screening [17–19]. The association of NT thickening and abnormal DV flow with subsequent PA/IVS diagnosis suggests these markers warrant thorough fetal echocardiography. Notably, all early-diagnosed cases exhibited the characteristic 3VT view finding of retrograde DA flow - a potentially pathognomonic feature when combined with pulmonary artery-aorta diameter discrepancy. However, technical limitations persist due to fetal size and positional factors, emphasizing the need for expert-level imaging.

Our stratification system, based on RV development and TV-Z scores, revealed important prognostic correlations. The inverse relationship between RV hypoplasia

**Table 3** Fetal conditions in PA/IVS with VCAC

| Case | Gestational week | TV-Z | Fetal heart | Combined extracardiac anomalies | Outcome |
|---|---|---|---|---|---|
| 1 | 13w | -4.19 | PA/IVS, VCAC, PE | NT thickening, single umbilical artery | Termination |
| 2 | 12w4d | -2.69 | PA/IVS, VCAC | reversed A-wave of the DV | Termination |
| 3 | 28w3d | -0.82 | PA/IVS, VCAC | / | Termination |
| 4 | 23w0d | -2.92 | PA/IVS, VCAC | / | Termination |
| 5 | 20w5d | -2.54 | PA/IVS, VCAC | / | Termination |
| 6 | 19w0d | -2.61 | PA/IVS, VCAC | / | Termination |
| 7 | 18w0d | -7.45 | PA/IVS, VCAC | / | Selective reduction of pregnancy |
| 8 | 24w2d | -0.29 | PA/IVS, VCAC | / | Termination |
| 9 | 22w4d | -6.13 | PA/IVS, VCAC, PE | / | Termination |
| 10 | 22w2d | -3.27 | PA/IVS, VCAC | / | Termination |
| 11 | 22w2d | -5.05 | PA/IVS, VCAC | / | Termination |
| 12 | 23w4d | -3.16 | PA/IVS, VCAC | / | Parturition, right coronary artery atresia by CTA, death. |
| 13 | 25w0d | -2.58 | PA/IVS, VCAC | reversed A-wave of the DV | Termination |
| 14 | 21w4d | -7.41 | PA/IVS, VCAC | / | Termination |
| 15 | 23w6d | 0.19 | PA/IVS, VCAC | ascites, reversed A-wave of the DV | Termination |
| 16 | 34w1d | -1.07 | PA/IVS, VCAC, PE | / | Termination |
| 17 | 24w0d | -3.5 | PA/IVS, VCAC, restrictive foramen ovale | reversed A-wave of the DV | Termination |
| 18 | 24w0d | -3.03 | PA/IVS, VCAC, PE | / | Termination |
| 19 | 21w4d | -4.29 | PA/IVS, VCAC, mild stenosis of the mitral and aortic valves, PE | reversed A-wave of the DV | Termination |

severity and VCAC prevalence (Type I:13.8% vs. Type III:80%) supports existing theories about coronary compensation mechanisms in restricted forward flow states [20]. The high VCAC incidence (39.6%) in our series may reflect referral bias but equally highlights the critical need for detailed coronary evaluation in prenatal assessment. This is particularly relevant given the established mortality risk associated with RVDCC [21], though our study confirms the ongoing challenge of reliable prenatal identification [22].

The 14.6% twin pregnancy incidence exceeds general population expectations [23], with particularly notable progression dynamics in TTTS cases. The observed transition from PS to PA/IVS in both recipients and donors complicates traditional volume-load paradigms [24–26], suggesting multifactorial mechanisms including RAS-mediated afterload effects and circular shunt physiology [27]. These findings reinforce current recommendations for intensified surveillance in monochorionic twins with hemodynamic imbalance.

While chromosomal abnormalities were rare (one 21q22.3 deletion), the potential association with microdeletion syndromes justifies our recommendation for comprehensive genetic testing [28, 29]. The 6.3% EA co-occurrence rate exceeds some literature reports [30], possibly reflecting improved detection through advanced imaging. The frequent DV flow abnormalities (12.5%) support their role as secondary markers of right heart dysfunction [31].

Clinical management implications emerge clearly from our outcomes data. The 86.4% termination rate for mid-late diagnoses reflects both disease severity and parental counseling realities. The single successful valvuloplasty case demonstrates potential for intervention in carefully selected membranous atresia, while the 80% mortality among live births underscores the critical importance of timely postnatal care coordination. The autopsy findings of fixed RVOT obstruction validate our imaging criteria while highlighting persistent technical challenges in pulmonary valve characterization.

### Limitations

This study has several limitations. As a retrospective study with partially incomplete data, it could not fully evaluate the diagnostic utility of fetal echocardiography for PA/IVS. The heterogeneous pathologic anatomy and dynamic intrauterine progression of PA/IVS also challenge the accuracy of prenatal diagnosis. Additionally, this single-center study had a small sample size, a high induction rate, and limited autopsy data, which may introduce bias in the results.

Future directions should focus on three areas: (1) refinement of early predictive algorithms incorporating NT/DV/TR parameters, (2) development of standardized

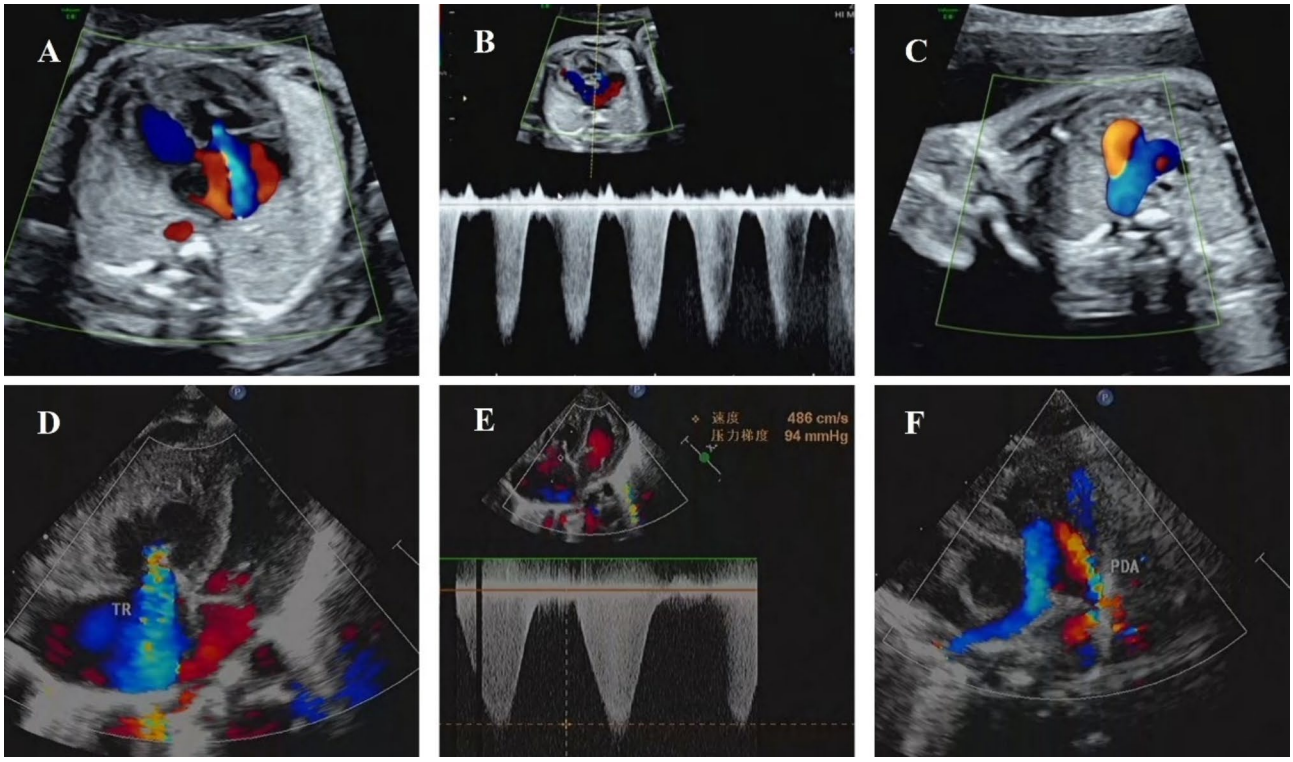


**Fig. 4** Prenatal and postnatal imaging findings of PA/IVS. (**A**) Prenatal ultrasound demonstrating severe regurgitation (large regurgitant jet); (**B**) Spectral doppler showing peak regurgitant velocity (416 cm/s). (**C**) Retrograde ductus arteriosus (DA) flow into pulmonary artery. (**D-F**). Postnatal echocardiography confirming prenatal findings with consistent anatomical and hemodynamic features

VCAC assessment protocols, and (3) multicenter studies to optimize selection criteria for fetal intervention. The progressive cases in our series particularly emphasize the need for dynamic risk assessment models that account for gestational evolution of disease severity.

## Conclusions

Fetal echocardiography is an effective tool for diagnosing PA/IVS. The diagnosis of PA/IVS can be made in early gestation, but it remains diagnostic challenge. Given the characteristic intrauterine progression of PA/IVS, sequential ultrasound examinations during the second and third trimesters are essential for monitoring disease progression and hemodynamic changes. Additionally, a comprehensive evaluation for potential associated intracardiac and extracardiac anomalies should be performed systematically throughout the pregnancy.

### Abbreviations

| | |
|---|---|
| PA/IVS | Pulmonary atresia with intact ventricular septum |
| CPS | Critical pulmonary stenosis |
| VCAC | Ventriculo-coronary artery communication |
| RVDCC | Right ventricular dependent coronary circulation |
| MCDA | Monochorionic diamniotic |
| MCMA | Monochorionic monoamniotic |
| DCDA | Dichorionic diamniotic |
| EA | Ebstein's anomaly |
| DA | Ductus arteriosus |
| DV | Ductus venosus |
| TV-Z | Z-score of the tricuspid valve |
| NT | Nuchal translucency |
| RVOTO | Right ventricular outflow tract obstruction |
| PA/VSD | Pulmonary atresia with ventricular septal defect |

### Author contributions

Q.W. and Z.W. designed and supervised the study. Q.H. were responsible for data collection, analysis and manuscript writing; H.T. worked on the literature review; W.L. and Q.W. were responsible for the design, implementation, and data analysis; T.D. and Z.Z. were responsible for some data collection. All authors contributed to the article and approved the submitted version.

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### Data availability

No datasets were generated or analysed during the current study.

## Declarations

### Ethics approval and consent to participate

The study was approved by the Ethics Committee of Fujian Maternity and Child Health Hospital (2024KY309). The patients/participants provided their written informed consent to participate in this study. Written informed consent was obtained from the individual(s) for the publication of any potentially identifiable images or data included in this article.

### Competing interests

The authors declare no competing interests.

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