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Prenatal Diagnosis Of Complex Congenital Heart Disease With Right Atrial Isomerism And Double Outlet Right Ventricle: A Combined Case Report

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Abstract

Background: *Congenital heart defects (CHDs) are the most frequently diagnosed prenatal anomalies, with complex subtypes such as double outlet right ventricle (DORV) and transposition of the great arteries (TGA) posing significant diagnostic and management challenges. The presence of such defects, particularly when associated with syndromes like right atrial isomerism, necessitates detailed fetal cardiac evaluation for early diagnosis and optimal perinatal care planning.*

Objective: *To assess fetal cardiac and abdominal anatomy through prenatal ultrasound and targeted fetal echocardiography in fetuses suspected of having complex congenital heart disease.*

Case Description: *The two cases of fetuses evaluated during the second trimester using transabdominal ultrasound and fetal echocardiography.*

Case 1: *A 21-week-3-day fetus underwent routine anomaly scanning. The fetus was structurally normal in terms of craniofacial, spinal, abdominal, and limb anatomy, with appropriate biometry and estimated fetal weight. However, cardiac assessment revealed DORV with D-TGA physiology and a small subpulmonic ventricular septal defect (~ 3 mm), with both great arteries arising from the right ventricle and lying in parallel orientation—findings consistent with an isolated conotruncal anomaly. No extracardiac or syndromic abnormalities were identified.*

Case 2: *A detailed fetal echocardiographic study in another fetus with suspected conotruncal anomaly revealed right atrial isomerism. Findings included bilateral right atrial appendages, absent spleen, right-sided stomach, midline liver, and left-sided aortic arch. Cardiac anomalies included a complete unbalanced atrioventricular septal defect (AVSD), DORV with D-TGA, subvalvular/valvular pulmonary stenosis, total anomalous pulmonary venous connection (TAPVC), and bilateral superior vena cavae—suggestive of heterotaxy syndrome with complex cardiac malformations.*

Conclusion: *These cases highlight the importance of prenatal ultrasound and echocardiography for early detection of complex CHDs, enabling timely counseling, planning, and improved outcomes in heterotaxy.*

Keywords; *Fetal echocardiography, double outlet right ventricle (DORV), transposition of great arteries (TGA), right atrial isomerism, heterotaxy syndrome, atrioventricular septal defect (AVSD), congenital heart defect (CHD).*

1. Introduction

Congenital heart defects (CHDs) are the most prevalent congenital anomalies, affecting approximately 8 in 1,000 live births, with complex variants significantly contributing to perinatal morbidity and mortality (1). The spectrum of CHDs ranges from isolated septal defects to complex malformations involving multiple cardiac and extracardiac structures. Among these, double outlet right ventricle (DORV) and transposition of the great arteries (TGA) are critical congenital heart diseases where both great arteries arise predominantly or entirely from the morphological right ventricle, often in association with ventricular septal defects (VSDs) and outflow tract obstruction (2, 3). These defects disrupt normal blood flow patterns and require prompt postnatal surgical

correction for survival. Another category of complex CHDs is heterotaxy syndrome, a disorder of abnormal left–right axis development leading to visceral and cardiac situs anomalies. Right atrial isomerism, also known as asplenia syndrome, is a severe form of heterotaxy characterized by bilateral right atrial morphology, midline liver, right-sided stomach, and frequently, absence of the spleen (4). It is strongly associated with major cardiac anomalies, including complete atrioventricular septal defects (AVSDs), pulmonary outflow tract obstruction, abnormal systemic venous return, and total or partial anomalous pulmonary venous connection (TAPVC/PAPVC) (5). Early and accurate prenatal diagnosis of such cardiac malformations is crucial for optimizing outcomes. Detailed second-trimester anomaly scans and targeted prenatal imaging, particularly the use of high-resolution fetal echocardiography, have significantly improved the detection and characterization of such complex cardiac defects in utero (6). Identifying these anomalies in utero facilitates appropriate parental counseling, delivery planning at tertiary care centers with pediatric cardiology and cardiac surgery support, and timely postnatal intervention (7).

In this case report, we present two prenatal cases: one with DORV and D-TGA, and another with right atrial isomerism associated with multiple complex intracardiac anomalies. These cases underscore the importance of detailed fetal cardiac assessment during second-trimester evaluation and the role of prenatal diagnosis in optimizing postnatal care for fetuses with life-threatening congenital heart diseases.

2. Case description

A combined presentation of two sequential prenatal evaluations outlines the complex cardiac and visceral findings in a single fetus, identified during routine second-trimester screening and subsequently confirmed by detailed fetal echocardiography. First pregnant woman underwent a routine second-trimester anomaly scan at 21 weeks and 3 days of gestation, calculated from her last menstrual period dated 23 September 2024. Transabdominal ultrasound, performed under good technical conditions, revealed a single live intrauterine fetus with biometric parameters appropriate for gestational age. However, in second pregnant woman a detailed fetal echocardiographic evaluation was performed on a fetus with a previously noted conotruncal anomaly during routine anomaly screening. The scan was carried out in the second trimester due to suspicious findings on a standard anomaly scan suggesting double outlet right ventricle (DORV) with D-transposition of the great arteries (D-TGA) and a subpulmonic ventricular septal defect (VSD). The objective was to further delineate fetal cardiac anatomy and assess associated thoracoabdominal structures.

3. Result

3.1. First case

Table 3.1.1 OB- 2/3 Trimester Scan Report

Category	Observation/Findings
Patient Details	**LMP:** 23/09/2024 **GA by LMP:** 21W 3D **EDD:** 30/06/2025
Scan Details	**Indication:** Anomaly Scan **Method:** Transabdominal USG **Technical Conditions:** Good
Fetal Status	Single live intrauterine fetus; Fetal activity and cardiac activity present; FHR: ~151 bpm
Presentation	Variable
Placenta	Anterior; Grade I
Liquor	Adequate
Umbilical Cord	Three-vessel cord (2 arteries, 1 vein)

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Table 3.1.2. Anatomical Survey

System	Findings
Head	Midline falx seen; Lateral ventricles and posterior fossa normal; No intracranial lesion seen
Face	Normal coronal and profile views; Orbits, nose, and mouth appear normal
Spine	Longitudinal and transverse views normal; Vertebrae and spinal canal normal
Thorax/Lungs	Both lungs seen; No pleural effusion
Heart	Normal size, axis, and position; Situs solitus; IVC and SVC drain into RA; No effusions or rhythm abnormality
	RA→RV and LA→LV continuity present. RV mildly enlarged compared to LV
	Both pulmonary artery and aorta arise from RV; Parallel orientation; Aorta anterior and right to PA
	Small subpulmonic VSD (~3 mm)
Abdomen	Situs normal; Liver, stomach, and bowel appear normal; Bladder and kidneys normal
Extremities	All long bones appropriate for GA; Hands and feet appear normal
Uterine Arteries	Normal Doppler flow

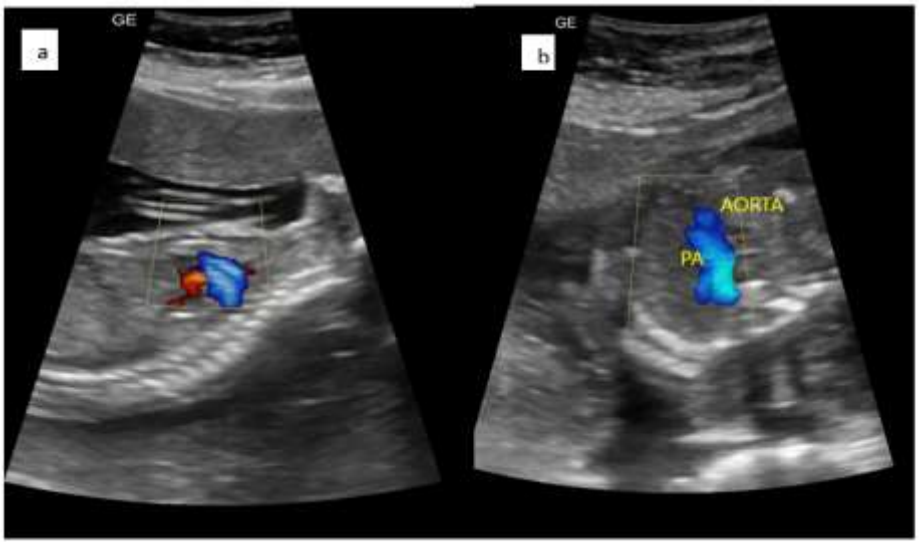
Table 3.1.3. Biometry

Parameter	Measurement	Gestational age
BPD	5.23cm	21W6D
HC	18.61cm	21W0D
AC	16.76cm	21W5D
FL	3.72cm	21W6D
EFW(BPD, HC, AC & FL)	446 +/- 67 grams	

A transabdominal obstetric ultrasound was performed at 21 weeks and 3 days of gestation for a routine anomaly scan. Above table 3.1, 3.2, and 3.3 presents a structured overview of fetal and maternal parameters as observed during the second-trimester scan. The fetus was viable, with appropriate activity and cardiac function, and the fetal heart rate was within normal limits (151 bpm). Fetal growth parameters, including biparietal diameter (BPD), head circumference (HC), abdominal circumference (AC), and femur length (FL), were consistent with the gestational age of approximately 21 weeks and 2 days, and the estimated fetal weight was 446 ± 67 grams. A detailed anatomical assessment revealed normal development of the fetal head, face, spine, extremities, and abdominal organs. The placenta was anterior with Grade I maturity, and amniotic fluid volume was within normal limits. The umbilical cord was three-vessel in structure, and uterine artery Doppler flow was normal bilaterally. Cardiac evaluation, however, revealed structural abnormalities. The heart was in normal position with situs solitus and correct venoatrial connections. The morphological right atrium was in continuity with the right ventricle, and the left atrium was continuous with the left ventricle. Notably, both the main pulmonary artery and ascending aorta arose from the morphological right ventricle and demonstrated a parallel course, with the aorta lying anterior and to the right of the pulmonary artery—findings indicative of double outlet right ventricle (DORV) with d-transposition of the great arteries (D-TGA)**.** Additionally, a small subpulmonic ventricular septal

defect (VSD) measuring approximately 3 mm was visualized. These observations are consistent with a conotruncal anomaly**.** No other extracardiac anomalies were detected. A follow-up scan was recommended at 28 weeks to monitor fetal growth and reassess the cardiac anatomy in greater detail.

Figure-1



Ultrasound images a) Mid sagittal section showing parallel orientation of main pulmonary artery and ascending aorta. B) Axial section showing ascending aorta is to the right of pulmonary artery.

3.2. Second case

Table 3.2.1. Fetal Echocardiography and Abdominal Findings

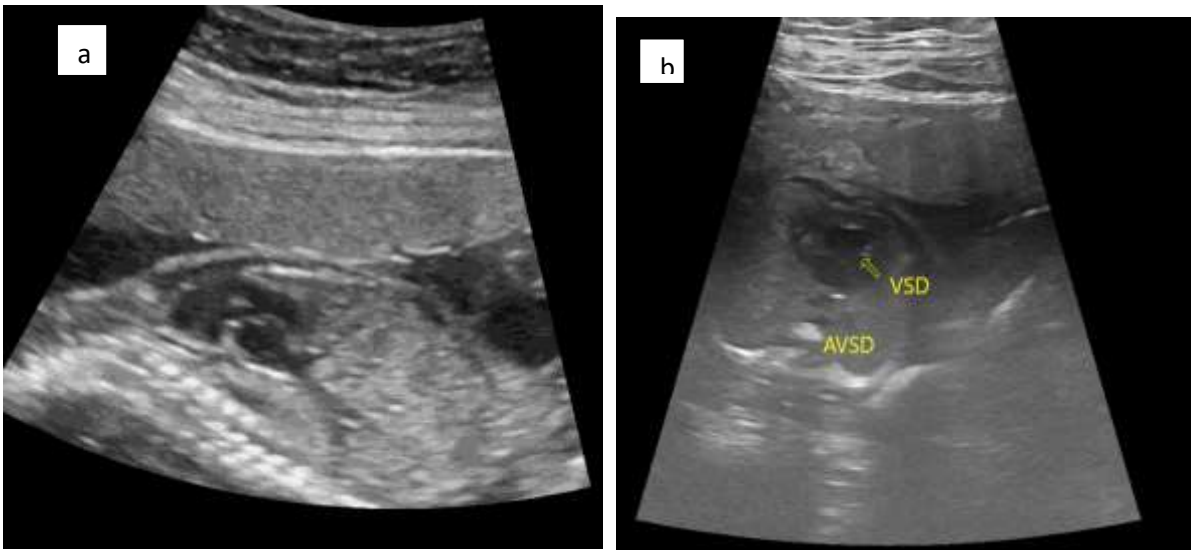
A detailed fetal echocardiographic evaluation was performed in the second trimester to assess suspected congenital heart disease and thoracoabdominal situs abnormalities. The ultrasound revealed multiple structural anomalies involving the cardiovascular system and visceral organs, consistent with a complex congenital syndrome. The observed findings are summarized in the table below.

System/Structure	Findings
Abdominal Situs & Vessels	IVC and abdominal aorta located on the left side, IVC anterior to the aorta
Liver	Enlarged, extending into right, midline, and left upper abdomen
Stomach	Positioned on the right side
Spleen	Not visualized
Heart Position & Size	Heart in midline, occupying ~1/3 of thoracic cavity; apex points left
Lungs	Both lungs appear normal in size and echogenicity
Atria	Prominent**;** both with pyramidal-shaped appendages (suggestive of right atrial isomerism)
Atrial Septum	Large ostium primum atrial septal defect (ASD)
Cardiac Crux	Not visualized
Ventricular Septum	Large membranous ventricular septal defect (VSD)
Right Ventricle	Enlarged**;** presence of moderator band

Left Ventricle	Small compared to right ventricle
Great Arteries	Both pulmonary artery and aorta arising from morphological right ventricle
Spatial Orientation of Vessels	Parallel arrangement; aorta anterior and right to pulmonary artery **(D-TGA)**
Pulmonary Stenosis	Subvalvular/valvular stenosis**;** narrowest diameter ~2.4 mm
Ductus Arteriosus	Prominent; flow from aortic arch to pulmonary artery
Aortic Arch	Enlarged**;** maximum calibre ~5.6 mm
Pulmonary Venous Return	Anomalous venous channel posterior to left atrium; no direct pulmonary vein drainage seen → Suspicious for TAPVC
SVC Anatomy	Double SVC (right and persistent left SVC)
Aortic Arch Side	Left-sided aortic arch
Effusions	None (no pericardial or pleural effusion)
Fetal Heart Rate	~140 bpm

These findings are highly suggestive of right atrial isomerism (asplenia syndrome) with associated unbalanced complete atrioventricular septal defect (AVSD)**,** double outlet right ventricle (DORV) with d-transposition of the great arteries (D-TGA)**,** subvalvular pulmonary stenosis**,** and a probable total anomalous pulmonary venous connection (TAPVC). The presence of a double SVC and midline heart position further support the diagnosis of visceral heterotaxy.

FIGURE-2:



Ultrasound images a) Midsagittal section showing main pulmonary artery and aorta arising from the morphological right ventricle. B) axial section showing membranous ventricular septal defect.

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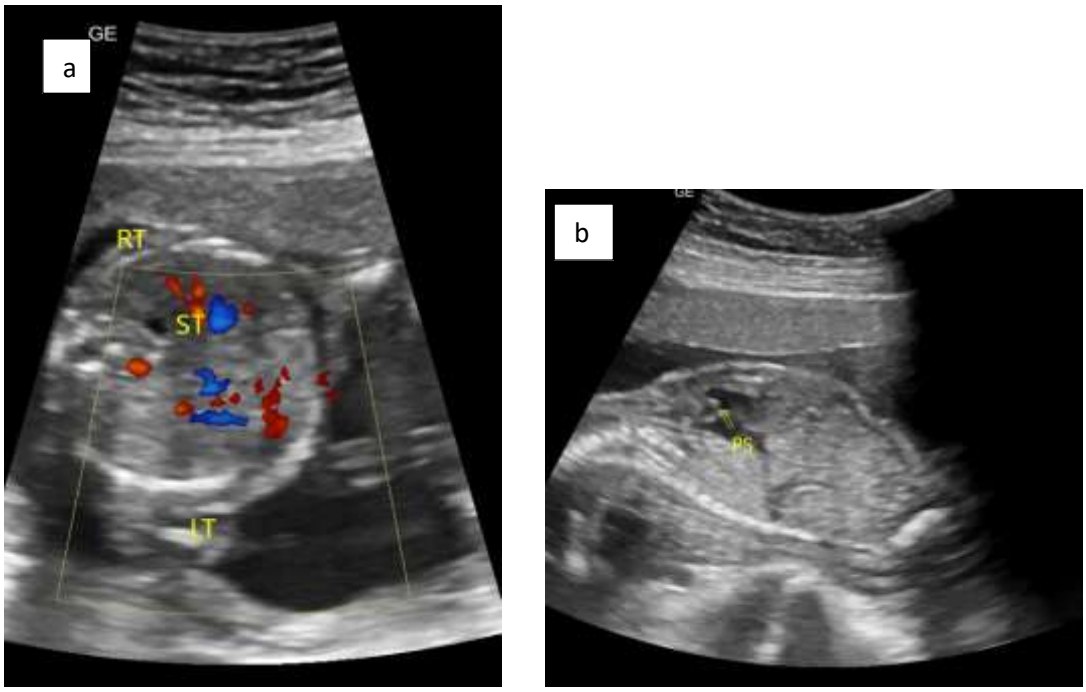
FIGURE-3



Ultrasound images a) axial section showing right sided stomach bubble b) sagittal section showing pulmonary stenosis with narrowed diameter measuring ~2.4mm.

4. Discussion

Congenital heart defects (CHDs) represent the most common form of congenital anomalies, with an estimated prevalence of 0.8% to 1.2% among live births globally, and can range from isolated simple lesions to complex malformations involving multiple cardiac and extracardiac structures (8). Among the more severe and rare CHDs are conotruncal anomalies, such as double outlet right ventricle (DORV) and transposition of the great arteries (TGA), which may occur in isolation or as part of syndromic associations like heterotaxy syndrome. The present case series highlights two distinct fetal presentations of DORV, one in isolation (Case 1) and the other in the context of right atrial isomerism (Case 2), providing insight into the variable phenotypic spectrum and prognostic implications.

Case Comparison and Significance

In Case 1, a structurally abnormal heart was identified in an otherwise normally sited fetus with concordant abdominal and thoracic organ arrangement. The key finding was DORV with d-transposition of the great arteries (D-TGA) and a subpulmonic VSD**.** Despite the presence of this conotruncal defect, other fetal anatomical systems appeared normal, and there were no markers suggestive of syndromic associations or chromosomal abnormalities. This form of DORV is classified as DORV with subpulmonic VSD and TGA physiology, where the aorta arises anteriorly and to the right of the pulmonary artery, and both vessels emerge from the right ventricle in a parallel course (9). Such cases may be amenable to postnatal surgical correction (e.g., arterial switch or Rastelli-type procedures), with generally better prognoses when unassociated with other anomalies (10).

In contrast, Case 2 represents a more complex cardiac and extracardiac malformation involving right isomerism (asplenia syndrome)**.** This condition is characterized by abnormal laterality of thoracoabdominal organs, often associated with complex cardiac malformations**,** including AVSDs**,** DORV**,** pulmonary outflow tract obstruction**,** and total anomalous pulmonary venous connection (TAPVC) (11). The findings of midline heart, bilateral pyramidal atrial appendages, absent spleen, right-sided stomach, and enlarged liver across the midline strongly support the diagnosis of right atrial isomerism. The echocardiographic presence of a complete AVSD**,** parallel

great vessels (indicative of D-TGA), and a suspicious anomalous pulmonary venous drainage further compound the severity of the diagnosis. Right isomerism is associated with a high incidence of complex CHDs and poor outcomes, particularly in the presence of pulmonary vein anomalies (12).

Embryological and Clinical Implications

DORV represents an abnormal conotruncal septation during cardiac development, leading to both the aorta and pulmonary artery arising predominantly from the right ventricle. This can be associated with a wide range of intracardiac and extracardiac anomalies, depending on the timing and severity of the embryological disruption (13). When DORV is seen in the setting of heterotaxy syndrome—as in Case 2—it often coexists with malpositioned abdominal organs, abnormal systemic venous return, and AVSDs due to failure of normal left-right patterning during early embryogenesis (14).

The detection of these anomalies prenatally has important clinical utility. Early diagnosis enables timely referral to tertiary care centers, multidisciplinary counseling, and planning for delivery at institutions equipped for neonatal cardiac intervention. While isolated DORV may be surgically corrected with favorable long-term outcomes, syndromic forms like those associated with right isomerism often require staged surgical palliation, and are associated with higher perinatal morbidity and mortality (15).

Recommendations and Prognostic Considerations

In both cases, early fetal echocardiography proved crucial in detecting significant anomalies. For Case 1, a relatively better prognosis is anticipated with isolated conotruncal abnormality, and surgical repair options are viable. In Case 2, however, the prognosis is guarded due to the combination of AVSD, DORV with D-TGA physiology, pulmonary stenosis, and suspected TAPVC. Long-term survival in right isomerism depends on the severity of pulmonary outflow obstruction and the feasibility of achieving functional circulation through palliative or corrective procedures (16). Both cases underscore the importance of comprehensive fetal cardiac assessment and situs evaluation during anomaly scans. Referral for detailed fetal echocardiography, genetic counseling, and multidisciplinary planning are imperative to optimize outcomes for such high-risk pregnancies.

5. Clinical Significance

The presented cases underscore the vital role of prenatal ultrasound and fetal echocardiography in the early detection and characterization of complex congenital heart defects (CHDs) such as double outlet right ventricle (DORV) and its associated anomalies. Timely identification of DORV, particularly when coupled with anomalies like d-transposition of the great arteries (D-TGA), atrioventricular septal defects (AVSDs), heterotaxy syndromes, and pulmonary venous abnormalities, enables comprehensive risk stratification and perinatal planning.

These findings hold considerable clinical importance for several reasons:

- **Perinatal Management**: Recognizing the presence of complex CHDs prenatally facilitates planned delivery at tertiary centers equipped with pediatric cardiac surgery and neonatal intensive care facilities, which can significantly influence neonatal survival and outcomes.
- **Parental Counseling**: Accurate diagnosis allows for early multidisciplinary counseling, providing families with information about prognosis, potential interventions, surgical options, and long-term care implications.
- **Surgical Planning**: Understanding the anatomical complexity (e.g., presence of DORV with AVSD and TAPVC) helps guide timing and nature of surgical interventions, such as single-ventricle palliation versus biventricular repair.

- **Genetic Evaluation**: The association of cardiac anomalies with visceral heterotaxy or isomerism suggests a potential underlying genetic etiology, prompting evaluation for syndromic associations and guiding further prenatal genetic testing when indicated.

Ultimately, these cases highlight the indispensable role of second-trimester anomaly scans and the need for fetal echocardiography referral in suspected or confirmed cardiac anomalies, ensuring optimal outcomes through early diagnosis, tailored intervention, and coordinated care.

6. Conclusion

This case series highlights the variable presentations and prognostic implications of double outlet right ventricle (DORV) in the prenatal period. Case 1, representing an isolated conotruncal anomaly, demonstrated DORV with d-transposition of the great arteries (D-TGA) and subpulmonic VSD in an otherwise structurally normal fetus—suggesting a potentially favorable outcome with appropriate postnatal surgical management. In contrast, **Case 2** illustrated a more complex scenario involving right atrial isomerism (asplenia syndrome), unbalanced atrioventricular septal defect (AVSD), DORV with D-TGA, pulmonary stenosis, and suspected total anomalous pulmonary venous connection (TAPVC)—features associated with high perinatal risk and requiring staged surgical palliation. These cases underscore the critical importance of detailed fetal echocardiographic evaluation, especially when conotruncal anomalies are suspected on routine anomaly scans. Early and accurate diagnosis allows for timely multidisciplinary counseling, planning of delivery at specialized centers, and improved perinatal and surgical outcomes. The integration of situs assessment and recognition of heterotaxy syndromes is essential for comprehensive prenatal cardiac diagnosis and prognostication.

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