Case Report

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Prenatal Diagnosis of Congenitally Corrected Transposition of the Great Arteries (CCTGA): A Case Report

Nasir Hematian[1], Shirin Torabi[2], Sedigheh Hantoushzadeh[3], Maasoumeh Saleh[4*], Mahboubeh Saleh[5]

1. Department of Perinatology and Fetal Cardiology, Yas Hospital, Tehran University of Medical Sciences, Tehran, Iran.
2. Perinatologist, Fetal-Maternal Research Center, Tehran, Iran.
3. Department Perinatology, Imam Khomeini Hospital Complex, Tehran University of Medical Sciences, Tehran, Iran.
4. Department Fetal-Maternal Medicine, Shariati Hospital, Tehran University of Medical Sciences, Tehran, Iran.
5. Fasa University of Medical Sciences, Fars, Iran.

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ABSTRACT

CCTGA, also known as levo- or L-loop transposition (L-TGA), double discordance, or ventricular inversion, is a rare cardiovascular anomaly with inversion of the ventricles and great arteries. In this anomaly, the right atrium communicates with the morphologic left ventricle, which gives rise to the pulmonary artery, while the left atrium communicates with the morphologic right ventricle, which gives rise to the aorta. Thus, atrioventricular and ventriculoarterial discordance (double discordance) exist, and although the blood flows in the normal direction, it passes through the wrong ventricular chambers. It is a unique conotruncal anomaly, in which the four-chamber view is abnormal. It may be associated with other heart disorders. In most fetuses, TGA remains undiagnosed before birth. The diagnosis of TGA can be made by carefully and appropriately evaluating the anatomic locations of cardiac chambers and the connections between the atria, ventricles, and great arteries with high-resolution ultrasound. Patients with isolated CTGA generally present later in life with signs and symptoms related to either arrhythmias or heart failure. TGA is rarely associated with chromosomal abnormality and amniocentesis is usually not undertaken. We report a case of CTGA detected at 18 week's gestation on screening ultrasound.

*** Corresponding Author:**
Maasoumeh Saleh
Address: *Department Fetal-Maternal Medicine, Tehran University of Medical Sciences, Shariati Hospital,Tehran, Iran.*
E-mail: *salehmaadsoumeh@yahoo.com.*

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Introduction

TGA was first described by Mathew Baillie in 1797 [1]. Corrected transposition of the great arteries (CTGA) is a very rare congenital heart disease with a published incidence ranging from 0.02 to 0.07 per 1000 live births, according to <1 percent of congenital heart disease lesions [2-4]. The cause of CTGA is likely multifactorial. Limited data suggest that both environmental and genetic factors contribute to its pathogenesis [5, 6]. This anomaly arises from left (Levo) looping of the developing heart tube [7]. In CCTGA both ventricles of the heart reversed. Fortunately, the arteries are reversed too. Therefore, the heart actually ''corrects'' the abnormal development; accordingly, the name ''congenitally corrected transposition of the great arteries". However, CCTGA is a complex malformation in which the heart is far from being normal. It is more frequently associated with other cardiac anomalies, such as ventricular septal defect (VSD), left ventricular outflow tract obstruction, anomalies of the left-sided tricuspid valve, and complete heart block [8]. Approximately, one-half of the fetuses with TGA have associated VSD [9]. CTGA occurs as an isolated defects in <10 percent of affected patients. Because prenatal diagnosis of CTGA is challenging, most patients with CTGA present after delivery. The antenatal diagnosis of CTGA is difficult, especially in isolated cases.

This condition can remain asymptomatic for several years in individuals without associated cardiopathies. Over the time, the systemic ventricle can become insufficient due to pressure overload. The treatment of this condition depends on the clinical presentation and the associated defect. It can go from the clinical treatment with drugs, to the palliative surgical treatment (physiological or anatomical with the arterial replacement) [10]. We describe a case of CTGA with VSD diagnosed in utero.

Case presentation

A 37-year-old woman, Grvida 2, Abortion 1, with a gestational age of 18 weeks was referred to our center for fetal echocardiography due to abnormal three vessel view in fetal ultrasound. She had no history of medical disease or drug usage. She had a legal abortion due to Down syndrome in previous pregnancy. After that, the parents underwent genetic testing that the Robertsonian translocation (14;21) was detected in the mother. In this pregnancy, her NT ultrasound was normal and amniocentesis showed normal 46XY karyotype. The indication for amniocentesis in this case was t h e history of Down syndrome in previous pregnancy and maternal chromosomal translocation. In the performed fetal echocardiography, the followings were detected: situs solitus, ventricular inversion in four-chamber view (Fig. 1), altered anatomic

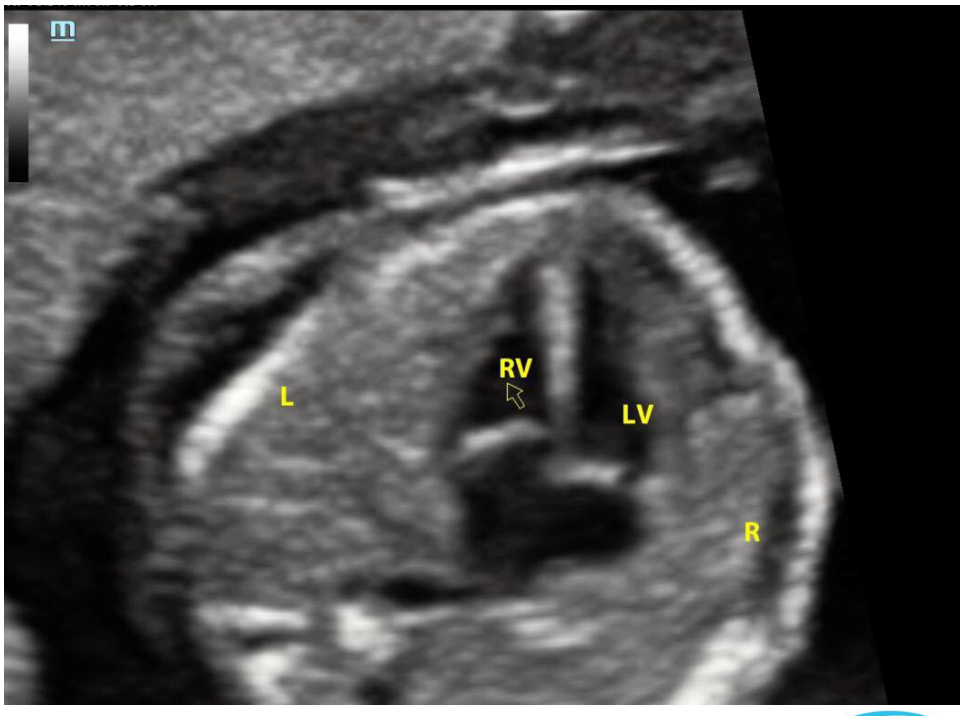

Fig. 1. Ventricular inversion in four-chamber view. (RV: right ventricle, LV: left ventricle, R: right, L: left).

sequence of vessels in three vessel view and ''I'' sign (boomerang sign) (Fig. 2, 3, 4), the parallel course of great arteries in outflow tract view (Fig. 5), aortic arch shift to the anterior of thorax, with a ductal arch in its posterior aspect (Fig. 6, 7) and VSD (Fig. 8). No other abnormalities were observed on ultrasound. All of these findings were in favor of diagnosing D-malposed CTGA.

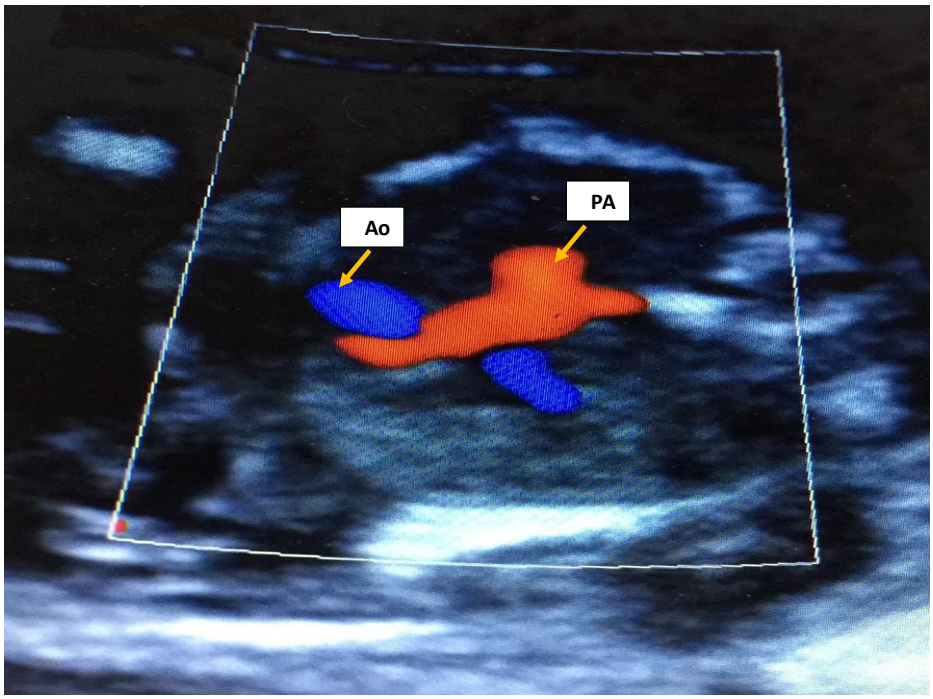

Fig. 2. Altered anatomic sequence of great arteries (intersection of aorta and pulmonary artery). (Ao: aorta, PA: pulmonary artery).

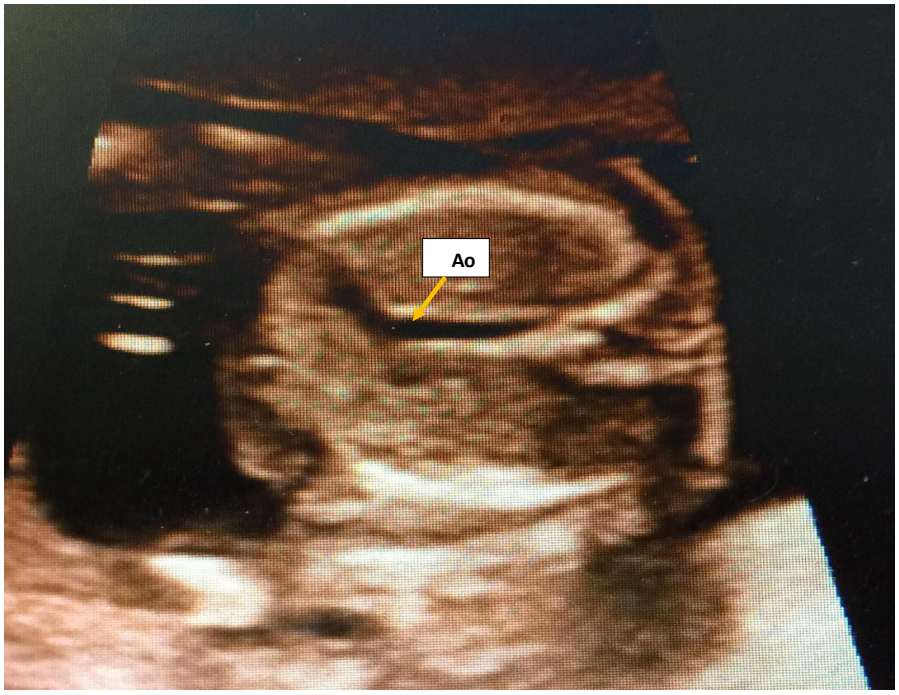

Fig. 3. ''I'' sign (boomerang sign) in three-vessel view. (Ao: aorta).

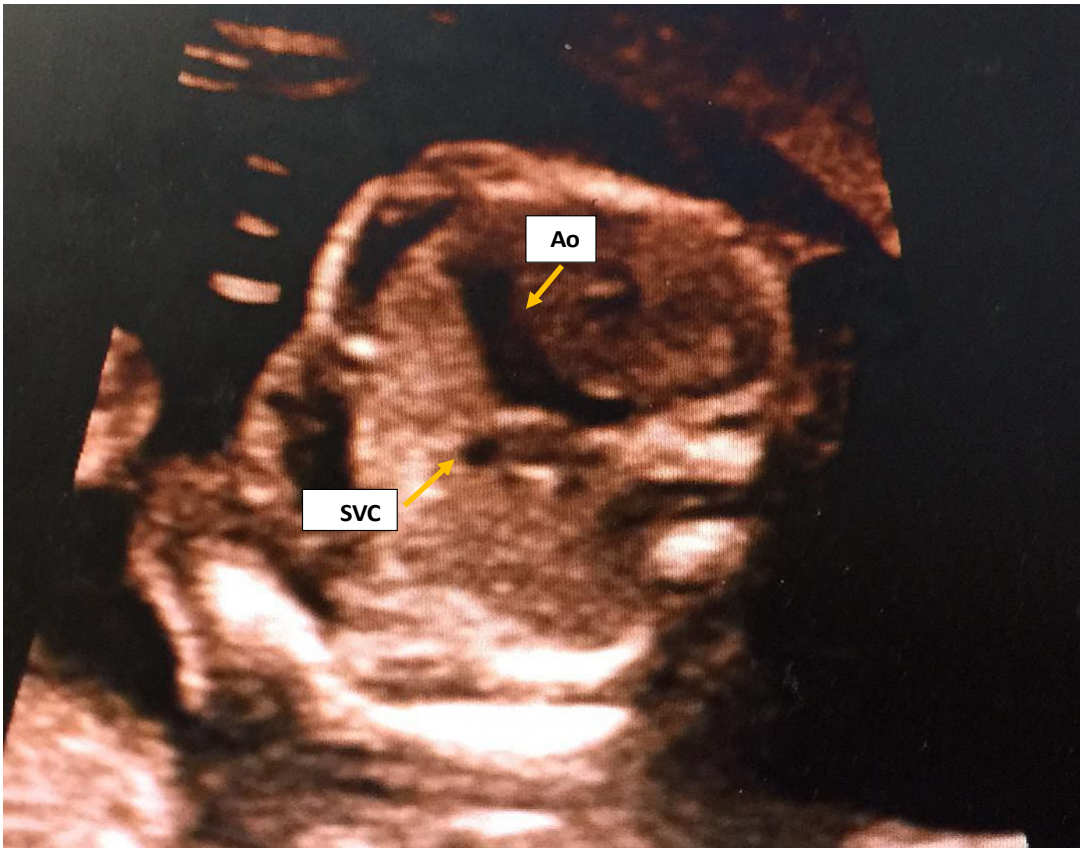

Fig. 4. ''I'' sign (boomerang sign) in three-vessel view.

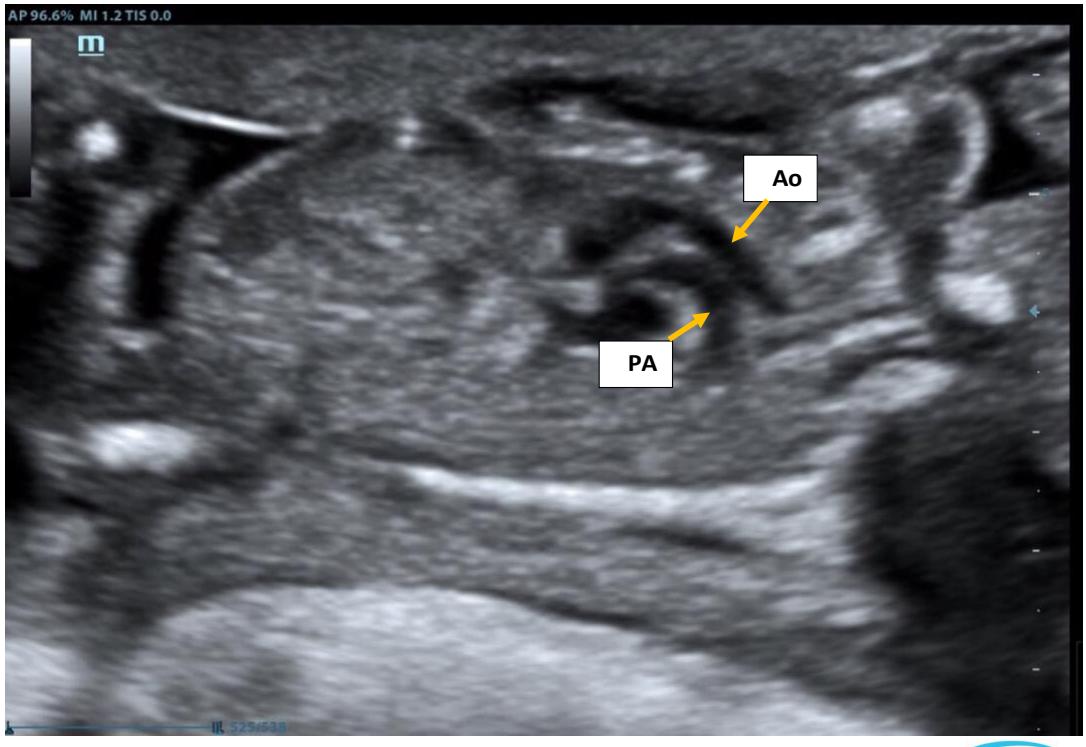

Fig. 5. The parallel course of great arteries in outflow tract view. (Ao: aorta, PA: pulmonary artery).

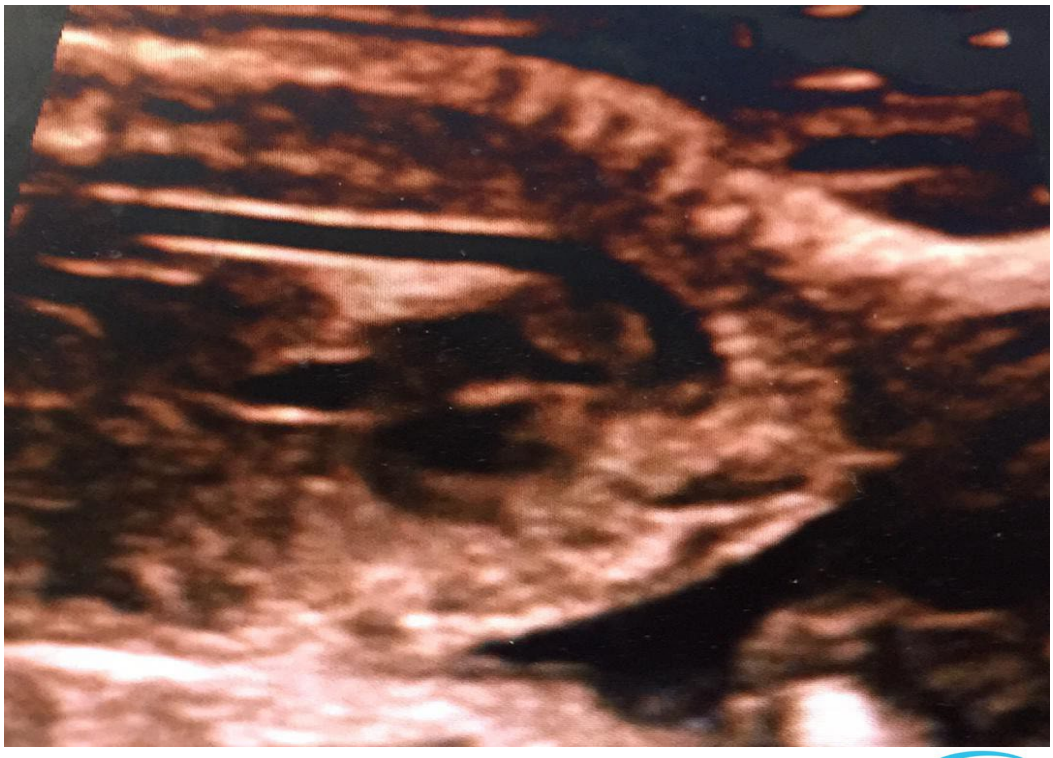

Fig. 6. The arch view. The aortic arch shifts to anterior of thorax with a ductal arch in its posterior aspect.

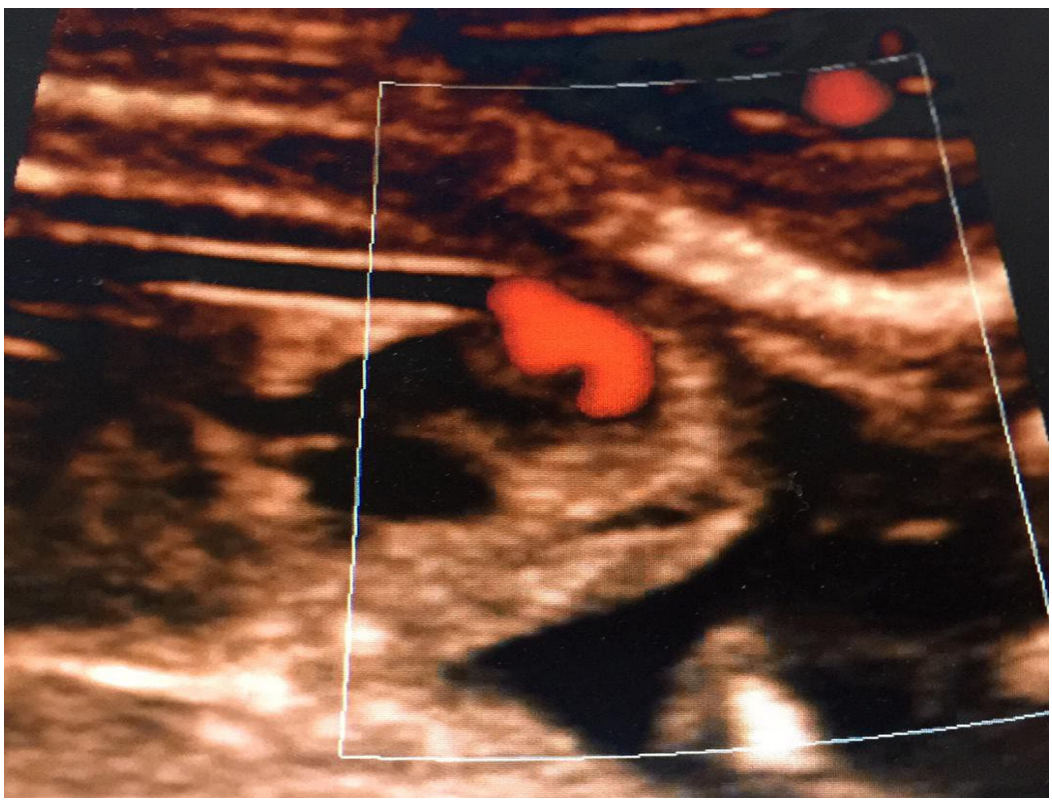

Fig. 7. The arch view in color Doppler.

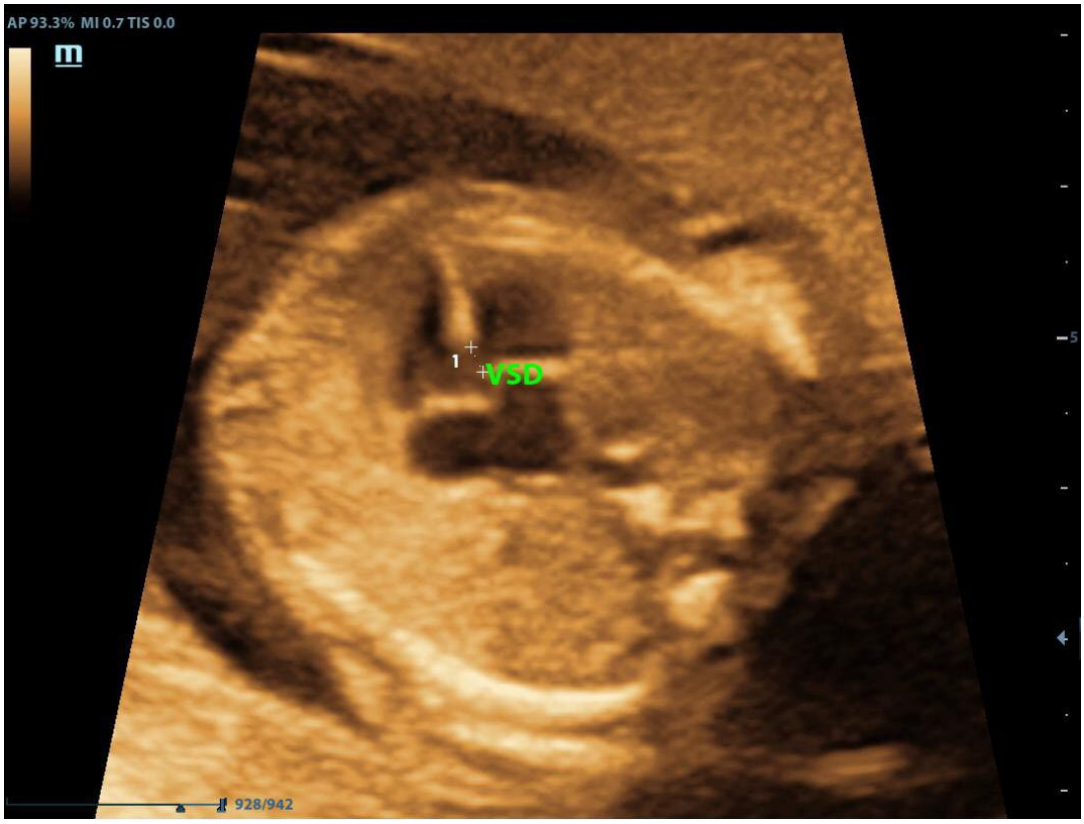

Fig. 8. VSD.

Discussion

In the normal heart, the left-side pumping chamber (left ventricle) sends blood to the entire body. The right-side ventricle pumps blood only a short distance to the lungs. There are two types of TGA that differ based on the vessels and heart chambers involved: Dextro-TGA (d-TGA) occurs when the position of the main pulmonary artery and aorta switched, L-TGA (CTGA) is a rarer condition in which not only the aorta and pulmonary arteries are switched but also the ventricles. In CCTGA, the heart twists abnormally during fetal development, and the ventricles are reversed: the stronger left ventricle pumps blood to the lungs and the weaker right ventricle has the harder chore of pumping blood to the entire body. CCTGA is a rare and complex congenital heart disease. There is no officially adopted classification for CTGA. Because the clinical presentation, prognosis, and treatment options are greatly influenced by the associated anomalies, it is useful to divide CTGA according to these anomalies into those with intact ventricular septum, those with VSD but without left ventricular outflow obstruction, and those with VSD and left ventricular obstruction. Our patient was in this category: CTGA with VSD but without left ventricular outflow obstruction. CTGA usually does not present with cyanosis unless there are associated cardiac defects, but they are at increased risk for heart failure in adulthood due to progressive decline in systemic right ventricular (RV) function. CCTGA may not be diagnosed on prenatal obstetric cardiac ultrasound, especially in the absence of other cardiac malformations (isolated CTGA) [11]. TGA is diagnosed prenatally in only 17% of neonates diagnosed after birth [9]. In affected fetuses without other cardiac defects, the four-chamber view could be interpreted as normal due to a lack of ventricular size discrepancy, as it is true in fetuses with dextro-TGA (D-TGA) [12]. Three-vessel view (3VV) is an important part of fetal cardiac imaging. In both types of TGA, the diagnosis is typically made by an experienced fetal ultrasonographer who correctly observe that the great arteries are parallel in the three-vessel view and do not cross as is expected in normal cardiac anatomy. The majority of cases are situs solitus [13]. Parallel vessels are evident in CTGA, but this sign is also presented in complete TGA (D- TGA). Because DTGA is a heart anomaly requiring major cardiac surgery in the postnatal period, it is important to differentiate between the entities prenatally [14]. TGA is more commonly associated with D-malposition of great

arteries where anterior aorta produces characteristic ''I'' sign in the 3VV in fetal heart imaging [15]. During fetal echocardiography, rightward convexity of the great vessel arising from the anterior ventricle and the ''I sign'' in 3VV have been considered as a pointer for TGA [16, 17]. In our case, these signs were noted. TGA is very rarely associated with genetic syndromes, such as turner, Noonan, Williams or Marfan syndrome, and in Down syndrome, it is virtually absent. The only genetic syndrome with a strong relation with TGA is heterotaxy. It is associated with fewer extracardiac anomalies (<10 percent) compared to other congenital heart defects [18]. No other anomalies were found in our case and fetal karyotype was normal. Due to the rarity of CCTGA, we still know little about the spectrum and the outcome of prenatally diagnosed cases [13]. Prenatal diagnosis is essential for better counseling of the parents and postnatal management. While the perinatal outcome is mostly favorable [19], our cases are at risk of postnatal development of right ventricle dysfunction, tricuspid valve regurgitation, complete heart block and congestive heart failure.

Conclusion

CCTGA is a cardiac anomaly that can be diagnosed prenatally with a good accuracy. It shows uneventful intrauterine course and has acceptable postnatal outcome with treatment options, especially in isolated cases.

Ethical Considerations

Consent

The consent was taken from the patient for the case report to be published.

Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.

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Authors' contributions

All authors equally contributed in preparing this article.

Conflict of interest

Authors declare that there is no conflict of interest.

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