

Received: 2019.10.19

Accepted: 2019.12.17

Available online: 2020.01.24

Published: 2020.02.29

# Late Presentation of a Congenitally Corrected Transposition of Great Arteries and Hemodynamically Balanced Ventricles Associated with a Large Ventricular Septal Defect and Severe Pulmonary Stenosis: A Case Report and a Multi-Imaging Approach

## Authors' Contribution:

Study Design A  
Data Collection B  
Statistical Analysis C  
Data Interpretation D  
Manuscript Preparation E  
Literature Search F  
Funds Collection G

ABCDEFG 1,2

ABCDEFG 2

ABCDEFG 2,3

ABCDEFG 2,4

ADG 2

**Khaled Elenizi****Rasha Alharthi****Anthony Matta****Abdulrahman Aljuayli****Michel Galinier**

1 Department of Internal Medicine, College of Medicine, Prince Sattam bin Abdulaziz University, Alkharj, Saudi Arabia

2 Cardiovascular Department, Rangueil Hospital, Toulouse, France

3 Faculty of Medicine, Holy Spirit University of Kaslik, Kaslik, Lebanon

4 Department of Surgery, College of Medicine, Al Imam Mohammad ibn Saud University, Riyadh, Saudi Arabia

**Corresponding Author:** Khaled Elenizi, e-mail: [kelenizi@yahoo.com](mailto:kelenizi@yahoo.com)**Conflict of interest:** None declared

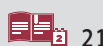
**Patient:** Male, 40-year-old  
**Final Diagnosis:** ccTGA  
**Symptoms:** Dyspnea  
**Medication:** —  
**Clinical Procedure:** —  
**Specialty:** Cardiology

**Objective:** Congenital defects/diseases**Background:** Congenitally corrected transposition of great arteries (ccTGA) represents a distinct rare group of congenital heart diseases. Survival of unoperated ccTGA in the presence of large ventricular septal defect (VSD) is exceptional. Furthermore, late presentation of such patients in the absence of severe pulmonary hypertension or severe systemic right ventricle dysfunction is unusual.**Case Report:** We report a rare late presentation of ccTGA associated with large VSD in the absence of severe pulmonary hypertension or systemic ventricle dysfunction. An associated severe pulmonary valve stenosis maintained a balanced and stable condition up to the fourth decade of life. The patient has also dextrocardia, which is an unusual association. The diagnosis was reached using multimodality imaging including transthoracic echocardiogram (TTE), transesophageal echocardiography (TEE), cardiac magnetic resonance imaging (CMR), and cardiac computed tomography (cardiac CT).**Conclusions:** The presence of pulmonary stenosis can provide a physiological protection that avoids unnecessary surgical correction of large VSD in ccTGA patients. However, such a decision should be made on an individual basis and following a careful anatomical and functional evaluation.**MeSH Keywords:** Cardiac Electrophysiology • Congenital Abnormalities • Transposition of Great Vessels**Abbreviations:** ccTGA – congenitally corrected transposition of the great arteries; LV – left ventricle; RV – right ventricle; LA – left atrium; RA – right atrium; TV – tricuspid valve; PA – pulmonary artery; TTE – transthoracic echocardiogram; TEE – transesophageal echocardiogram; VSD – ventricular septal defect; TR – tricuspid regurgitation; MR – mitral regurgitation; AV – atrioventricular; VA – ventriculo-arterial; CMR – cardiac magnetic resonance; Cardiac CT – cardiac computed tomography; CHF – congestive heart failure; PS – pulmonary stenosis; PHT – pulmonary hypertension; NYHA – New York Heart AssociationFull-text PDF: <https://www.amjcaserep.com/abstract/index/idArt/920822>

996



8



21



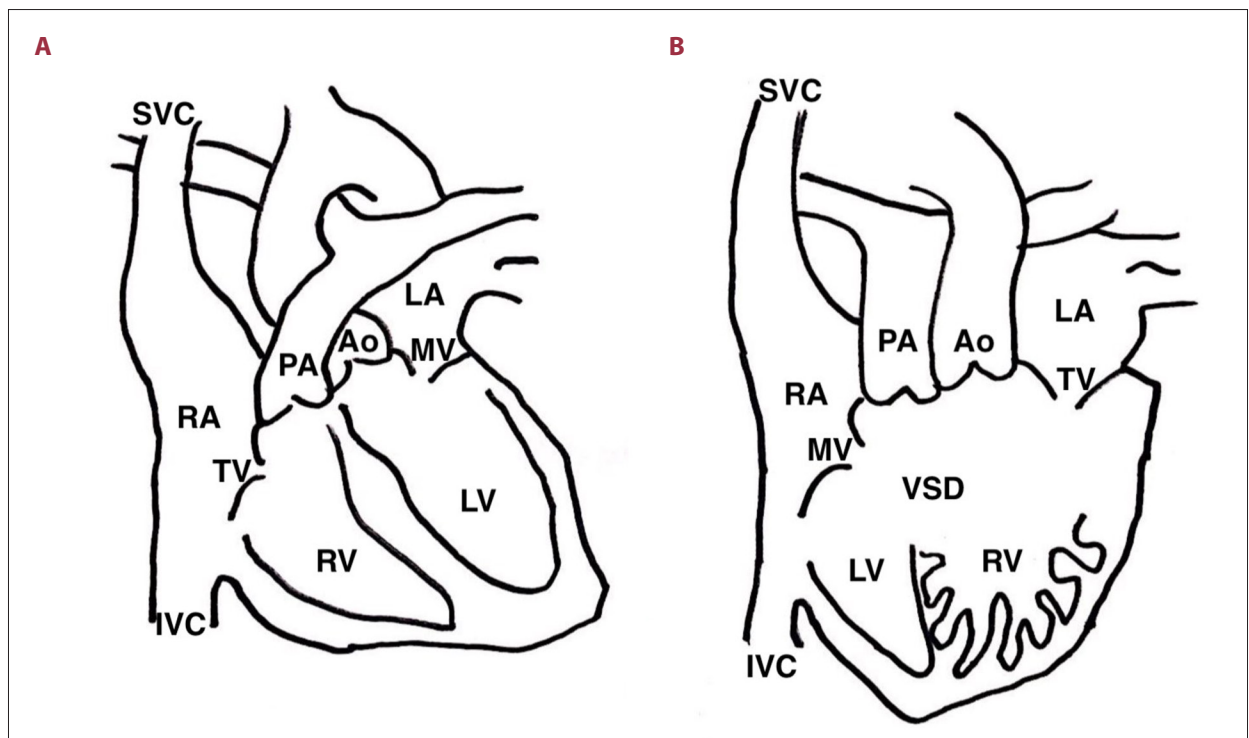
## Background

ccTGA is a rare congenital heart disease that represents less than 1% of all congenital heart diseases [1,2]. This malformation is characterized by atrioventricular (AV) discordance and ventriculo-arterial (VA) discordance (Figure 1). Various other terms anatomically describe this malformation: L-transposition, double discordance, and ventricular inversion.

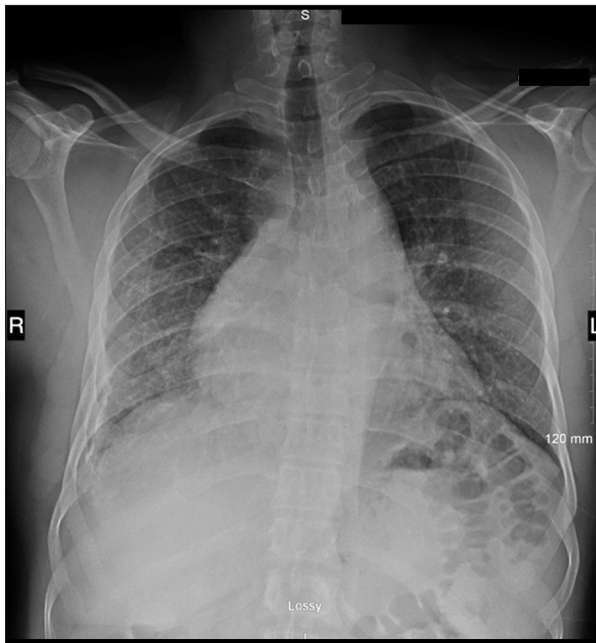
The term “congenitally corrected” is used to differentiate this condition from complete transposition (dextro-TGA) in which there is VA discordance but AV concordance. Eighty percent of ccTGA patients have associated anomalies, mostly VSD, PS, and both [3,4]. Others include Ebstein’s anomaly, tricuspid regurgitation, and, less commonly, mitral regurgitation [5–21]; less than 1% have no associated lesions [6]. Isolated ccTGA is considered physiologically corrected as passage of oxygenated and deoxygenated blood to the lungs and systemic circulation remains normal. However, the systemic morphologically RV is in contact with the systemic circulation, resulting in mechanical difficulty that could appear with aging [7].

## Case Report

A 40-year-old male, farmer, refugee, not known to have any medical illnesses, presented with NYHA class II dyspnea. Cardiac auscultation found a pan-systolic murmur, lungs were clear to auscultation, and oxygen saturation 94% on room air. Lab workup include hemoglobin 19.6 g, hematocrit 53%, platelets 219 g/l, WBC 6.6 g/l, normal electrolytes, NT-proBNP 35 pg/ml, normal renal function, and normal liver function. CXR showed clear lung fields, cardiomegaly, and dextrocardia (Figure 2). ECG revealed a QRS reversal pattern with dominant R wave but no Q wave in V6 and a first-degree AV block (Figure 3). TTE found atrioventricular and ventriculo-arterial discordances, lower displacement of the tricuspid valve indicative of Ebstein’s anomaly, and severe pulmonary stenosis with the pulmonary artery (PA) displaced to the right side of the aorta (Figure 4). TEE showed a pulmonary stenosis with malposition of the PA at the right side of the ascending aorta (Figure 5). Cardiac MRI showed a preserved bi-ventricular function and large VSD ( $\approx 3$  cm), RV hypertrophy, and dilation at indexed RV volume of 129.4 ml/m<sup>2</sup>, increased RV trabeculations, PA dilation at 46 mm, no late gadolinium enhancement, and nonsignificant mitral regurgitation (Figures 6, 7). A CT scan showed dextrocardia with double discordance (Figure 8). A right heart catheter showed pulmonary protection with right atrial pressure



**Figure 1.** Drawing of ccTGA with large VSD and dextrocardia adapted to our case. The RV is identified by virtue of trabeculations (A), normal heart for comparison (B). LV – left ventricle; RV – right ventricle; LA – left atrium; RA – right atrium; TV – tricuspid valve; Ao – aorta; PA – pulmonary artery; VSD – ventricular septal defect; SVC – superior vena cava; IVC – inferior vena cava; ccTGA – congenitally corrected transposition of the great arteries.



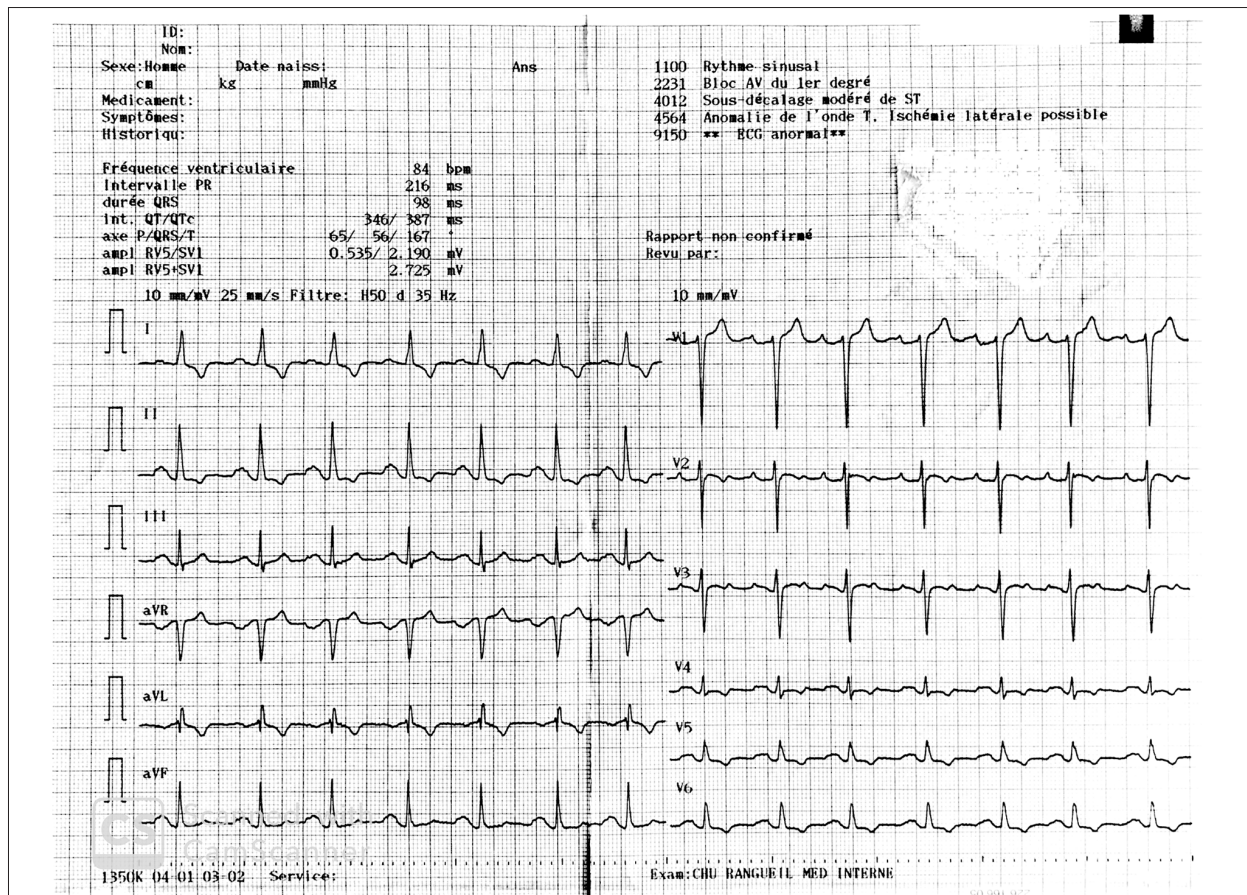
**Figure 2.** Chest x-ray shows dextrocardia and absent convexities of the descending aortic knob and pulmonary trunk on the left side.

7 mmHg, pulmonary capillary wedge pressure (PCWP) 9 mmHg, left ventricular pressure (LVP) 130/12 mmHg, pulmonary artery pressure (PAP) 25/15 (8) mmHg, PA partial pressure of oxygen (pO<sub>2</sub>) 46.2 mmHg, and superior vena cava (SVC) pO<sub>2</sub> 34.7 mmHg. Stress testing showed low functional capacity, with maximal oxygen uptake at 13.3 ml/min/kg.

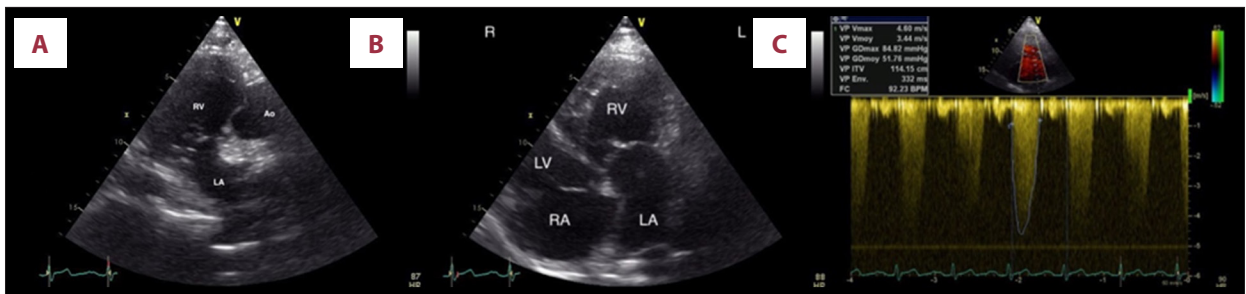
Surgical correction was not indicated in the presence of a hemodynamic balance, absence of significant tricuspid regurgitation, stable symptomatology, and pulmonary protection, despite the large VSD.

## Discussion

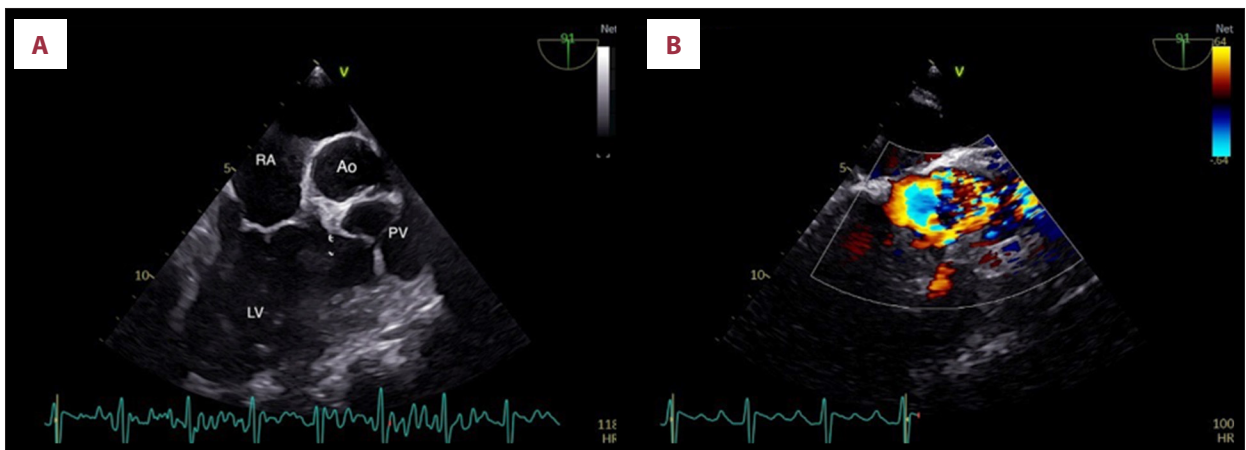
The clinical presentation of ccTGA depends on the presence or absence of associated cardiac anomalies. In the pediatric population, developing cyanosis and heart failure is related to the severity of associated malformations like a significant VSD or pulmonary stenosis, which might need surgical correction. In the absence of associated defects, which is uncommon, the RV can maintain its function as the systemic pump at a “normal”



**Figure 3.** ECG shows QRS reversal pattern with dominant R wave but no Q wave in V6 and first-degree AV block (PR 240 ms).



**Figure 4.** Transthoracic echocardiography in long-axis view showing RV morphology in left-sided systemic ventricle (A), 4-chamber view showing ventricular inversion. The RV has obviously more trabeculation and a more apically displaced tricuspid valve than that of the mitral valve (B). Echo Doppler shows severe pulmonary stenosis (C).



**Figure 5.** (A, B) Transesophageal echocardiography shows LV morphology connected to the PA with pulmonary valve stenosis and the aorta located anterior and displaced to the left.

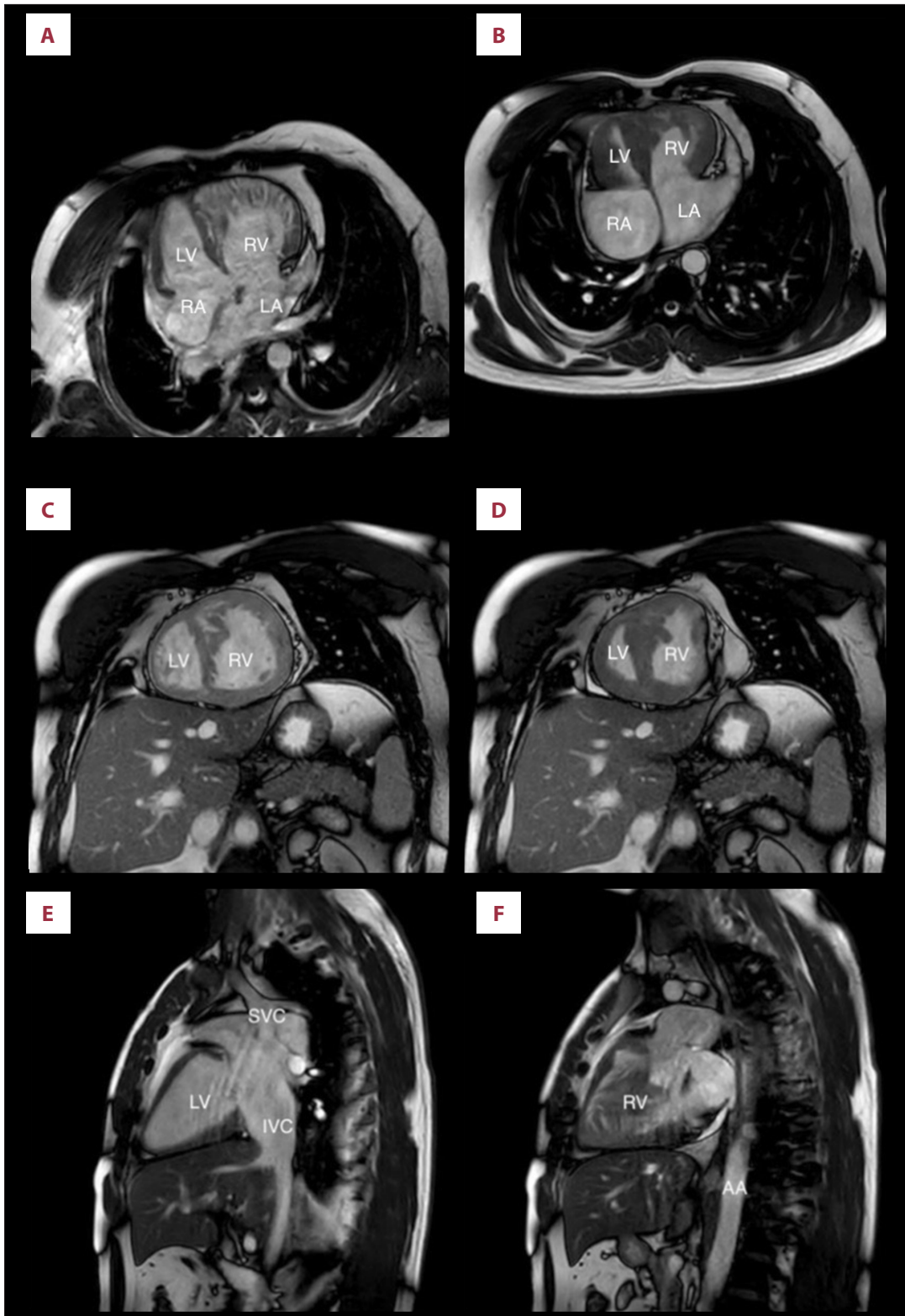
level into adult life, but it tends to deteriorate gradually after the second decade of life [8].

RV dysfunction is common in middle-aged adults with cCTGA [9], and individuals without associated cardiac defects may survive over 50 years [10,11]. However, survival of unoperated patients until the fourth decade in the presence of large VSD is exceptional. Several multicentric studies found an increasing occurrence of heart failure with age. In the fourth decade of life, half of cCTGA patients with associated lesions and one-third of those without associated lesions present with dysfunction of the systemic RV [12,13].

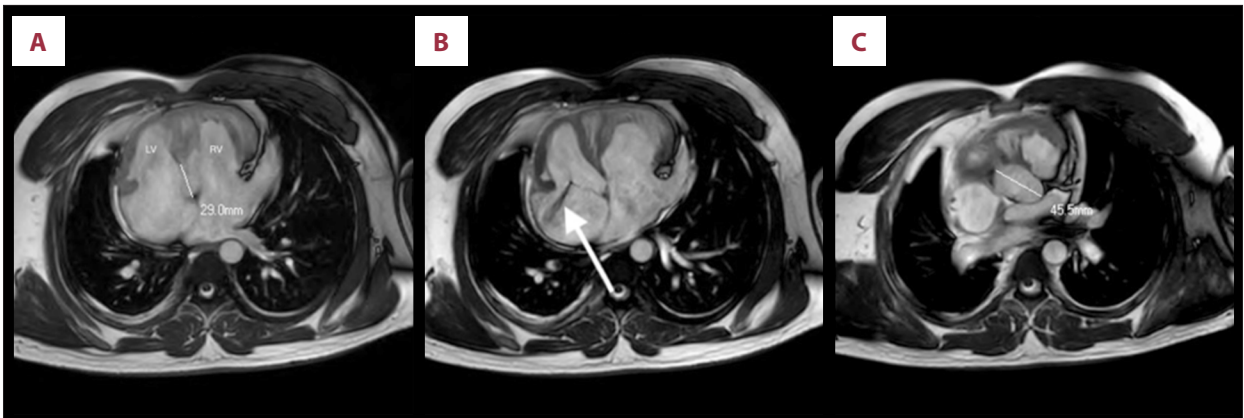
Large VSDs are usually associated with pulmonary hypertension (PHT). However, we found pulmonary protection in our case due to pulmonary valve stenosis. The increased pressure in LV helps maintain TV leaflets coaptation by compressing the RV [14] and the same applies for pulmonary artery banding. This reduces the risk of developing severe tricuspid regurgitation (TR), which is strongly associated with RV dysfunction and CHF in cCTGA patients [3]. Whereas it is extremely difficult to determine whether TR plays a major or a minor role [3], tricuspid valve repair is one of the surgical approaches that

showed better survival for those with significant tricuspid regurgitation [15].

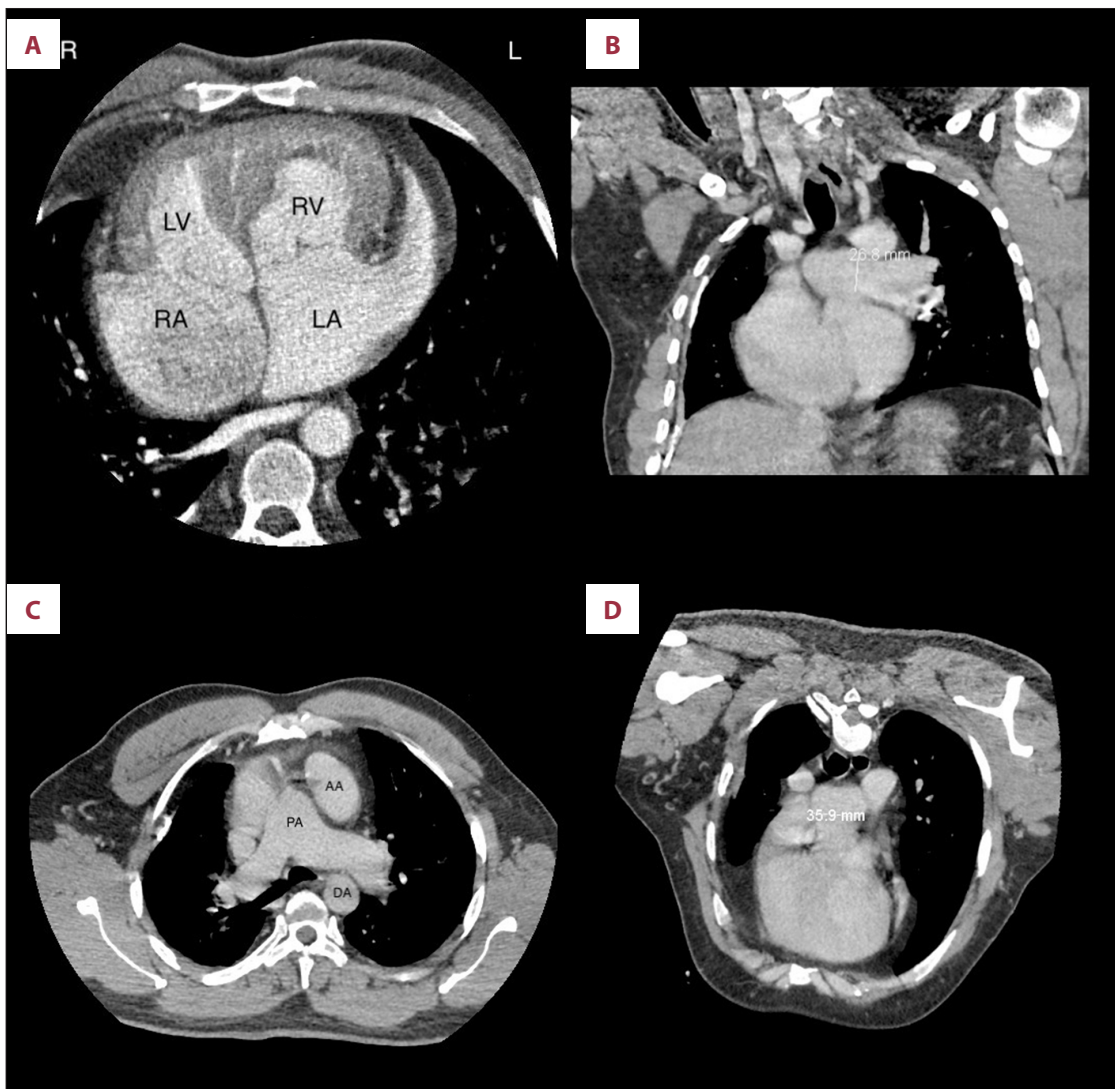
From an electrophysiological point of view, patients with cCTGA usually have a second AV node positioned anteriorly, which gives rise to an elongated AV bundle [16]. As a result, those patients are prone to have varying degrees of conduction abnormalities and re-entry tachycardia that manifest at any age from fetal life to late adulthood [17]. Diagnosis of cCTGA leading to complete heart block is well known [18]. The right bundle adheres to the RV and the left bundle adheres to the LV. Septal activation occurs in the opposite direction, from right to left [19]. Therefore, the ECG shows a mirror image pattern in the precordial leads, leading to a reversal of the normal QRS morphology, with Q waves in V1 and a dominant R wave but no Q wave in V6. In our patient, this mirror image pattern was partially evident with dominant R wave but no Q wave in V6 and absence of Q waves in V1, probably related to the RV hypertrophy. P waves and QRS axis vary depending on the atrial arrangement and location of the AV node and proximal conducting bundles [20]. Another anatomical finding in our case was the associated dextrocardia, which is not a common association [21].



**Figure 6.** Cardiac MRI shows ventricular inversion with the RV on the left side differentiated by trabeculations and morphology in diastole (A) in systole (B), short axis in diastole (C) in systole (D), sagittal plane shows LV morphology connected to IVC and SVC (E), RV morphology connected to the aorta (F). AA – abdominal aorta; SVC – superior vena cava; LA – left atrium; LV – left ventricle; RA – right atrium; RV – right ventricle.



**Figure 7.** Cardiac MRI 4-chamber view shows VSD of 29 mm (A), mitral regurgitation (B), coronal view shows pulmonary trunk dilation of 45 mm (C).



**Figure 8.** Chest CT scan shows RV by virtue of predominant apical trabeculations connected to the systemic circulation (A), ascending aorta laterally to the left and anterior of the pulmonary artery (B). Coronal plane CT scan shows dextrocardia with normal size left PA (C). Axial plane CT scan shows the pulmonary artery (D).

## Conclusions

Patients with ccTGA can live for years without diagnosis. The prognosis depends largely on the ability of the RV to adapt to the systemic circulation. Surgical correction is achieved mainly by reverting the ventricles, and its indication relies on a multimodality imaging approach and exhaustive hemodynamic assessment. The presence of pulmonary stenosis can provide a physiological protection that avoids unnecessary surgical correction.

## References:

1. Connelly MS, Liu PP, Williams WG et al: Congenitally corrected transposition of the great arteries in the adult: Functional status and complications. *J Am Coll Cardiol*, 1996; 27(5): 1238–43
2. Graf M, Zaczekiewicz M, Torzewski J, Zimmermann O: Atrial fibrillation-induced cardiac shock: First manifestation of a congenitally corrected transposition of the great arteries in a 45-year-old man. *Case Rep Cardiol*, 2012; 2012: 126764
3. Alghamdi AA, McCrindle BW, Van Arsdell GS: Physiologic versus anatomic repair of congenitally corrected transposition of the great arteries: Meta-analysis of individual patient data. *Ann Thorac Surg*, 2006; 81(4): 1529–35
4. Kantarci M, Koplay M, Bayraktutan U et al: Congenitally corrected transposition of the great arteries: MDCT angiography findings and interpretation of complex coronary anatomy. *Int J Cardiovasc Imaging*, 2007; 23(3): 405–10
5. Wallis GA, Debich-Spicer D, Anderson RH: Congenitally corrected transposition. *Orphanet J Rare Dis*, 2011; 6: 22
6. Connolly HM, Miranda WR, Egbe AC, Warnes CA: Management of the adult patient with congenitally corrected transposition: Challenges and uncertainties. *Semin Thorac Cardiovasc Surg Pediatr Card Surg Annu*, 2019; 22: 61–65
7. Prieto LR, Hordof AJ, Secic M et al: Progressive tricuspid valve disease in patients with congenitally corrected transposition of the great arteries. *Circulation*, 1998; 98(10): 997–1005
8. Grolleau R, Baissus C, Puech P: Congenitally corrected transposition of the great vessels and preexcitation syndrome (apropos of 2 cases). *Arch Mal Coeur Vaiss*, 1977; 70: 69
9. Graham TP Jr., Bernard YD, Mellen BG et al: Long-term outcome in congenitally corrected transposition of the great arteries: A multi-institutional study. *J Am Coll Cardiol*, 2000; 36(1): 255–61
10. Ikeda U, Furuse A, Suzuki O et al: Long-term survival in aged patients with corrected transposition of the great arteries. *Chest*, 1992; 101: 1382–85
11. Cummings GR: Congenital corrected transposition of the great vessels without associated intracardiac anomalies. *Am J Cardiol*, 1962; 10: 605–14
12. Dimas AP, Moodie DS, Sterba R, Gill CC: Long-term function of the morphological right ventricle in adult patients with congenitally corrected transposition of the great arteries. *Am Heart J*, 1989; 118: 896
13. Kishon Y, Shem-Tov AA, Schneeweiss A, Neufeld HN: Congenitally corrected transposition of the great arteries without associated defects – a study of 10 patients. *Int J Cardiol*, 1983; 3: 112
14. Kral Kollars CA, Gelehrter S, Bove EL, Ensing G: Effects of morphologic left ventricular pressure on right ventricular geometry and tricuspid valve regurgitation in patients with congenitally corrected transposition of the great arteries. *Am J Cardiol*, 2010; 105: 735–39
15. Van Son JA, Danielson GK, Huhta JC et al: Late results of systemic atrioventricular valve replacement in corrected transposition. *J Thorac Cardiovasc Surg*, 1995; 109: 642–52; discussion 652–53
16. Anderson RH, Becker AE, Arnold R et al: The conducting tissues in congenitally corrected transposition. *Circulation*, 1974; 50: 911–23
17. Sharland G, Tingay R, Jones A, Simpson J: Atrioventricular and ventriculoarterial discordance (congenitally corrected transposition of the great arteries): Echocardiographic features, associations, and outcome in 34 fetuses. *Heart*, 2005; 91: 1453–58
18. Kafali G, Elsharshari H, Ozer S et al: Incidence of dysrhythmias in congenitally corrected transposition of the great arteries. *Turk J Pediatr*, 2002; 44: 219–23
19. Warnes CA: Transposition of the great arteries. *Circulation*, 2006; 114: 2699–709
20. Mah K, Friedberg MK: Congenitally corrected transposition of the great arteries: Situs solitus or inversus. *Circ Cardiovasc Imaging*, 2014; 7: 849–51
21. Hornung TS, Calder L: Congenitally corrected transposition of the great arteries. *Heart*, 2010; 96: 1154–61

## Acknowledgement

We would like to express our special thanks to Prince Sattam bin Abdulaziz University for support of this research.

## Conflict of interest

None.