Ann Med Res 2021;28(6):1263-5

Annals of Medical Research

DOI: 10.5455/annalsmedres.2020.11.1130

Case Report

A rare cause of dyspnea: Isolated congenitally corrected transposition of the great arteries

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Abstract

Corrected transposition of the great arteries is a rare pathology with an incidence of 1% among the congenital heart diseases. It is thought to have multifactorial inheritance. It is mostly seen in men. If there is no concomitant pathology, the blood flow is physiological and does not show symptoms until advanced ages. Life expectancy is close to normal in those with isolated pathology. Risk factors determining mortality include progressive right ventricular dysfunction, AV block, and severe tricuspid insufficiency. In this case, we present a CCTGA patient with dyspnea symptoms without any additional pathology.

Keywords: Congenital heart disease; dyspnea; echocardiography

INTRODUCTION

Congenitally corrected transposition of the great arteries (CCTGA) is a rare congenital heart disease. It is seen in 1%. CCTGA defines the physiologically corrected blood flow direction due to the combination of ventricular inversion (atrioventricular discordance) and ventriculoarterial discordance (1).

It is often accompanied by congenital heart defects. The most common accompanying pathologies include ventricular septal defect (VSD), pulmonary stenosis, dextrocardia, tricuspid valve anomalies (dysplasia, straddling, Ebstein). Sometimes, it can also be accompanied by hypoplastic ventricle, atrioventricular (AV) valve anomalies and complex cardiac defects with a number of VSDs (2). In 9-14.7% of CCTGA cases, there is no accompanying cardiac pathology (3). In isolated CCTGA, blood flow is physiological, and CCTGA does not show symptoms until advanced age and life expectancy is close to normal (1,2). In this article, we would like to present a case of CCTGA that was admitted to our outpatient clinic with dyspnea on exertion and that, albeit very rarely, should be considered in the differential diagnosis.

CASE REPORT

An 18-year-old female patient was admitted to the outpatient clinic with a complaint of dyspnea. The patient had effort dyspnea for the last 2 months and did not have any significant history. In the physical examination, blood pressure was 110/65 mmHg, and pulse was 64 beats/min and rhythmic. Cardiac examination revealed a 3/6 holosystolic murmur at the fifth intercostal space on the left during auscultation. Electrocardiographic evaluation revealed normal sinus rhythm, absence of R wave in lead V1, and absence of Q wave in left precordial leads (Figure 1). The transthoracic echocardiography demonstrated atrioventricular and ventriculoarterial discordance. While the pulmonary artery originated from the anatomical left ventricle located on the right, the aorta originated from the anatomic right ventricle located on the left (Figure 2). Hypertrophy and dilatation were observed in the systemic ventricle (right ventricle). Mild regurgitation was observed in the systemic AV valve (tricuspid valve). Systemic ventricle was global mild hypokinetic and the ejection fraction was 45%. No concomitant defect was observed. To confirm the diagnosis, cardiac computed tomography (CT) was requested as an advanced examination (Figure-3). The aorta was observed to be in front of the pulmonary artery. No tachycardia or bradycardia was detected in the 24-hour rhythm holter recording. Medical treatment was decided for the patient. ACE inhibitor was started and the patient was monitored.

Received: 19.11.2020 **Accepted:** 26.01.2021 **Available online:** 25.06.2021
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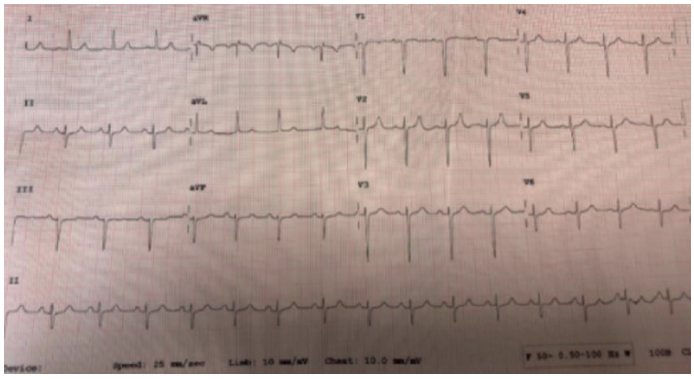

Figure 1. 12-lead electrocardiogram of the patient

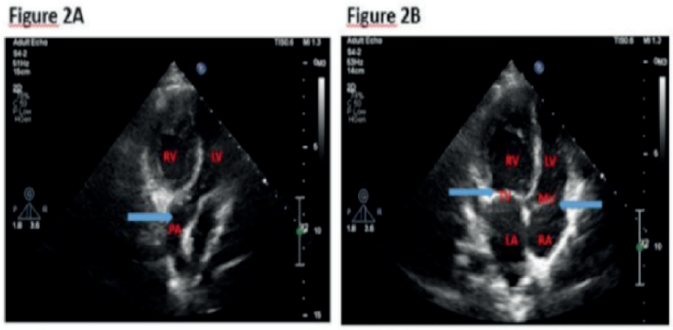

Figure 2. A: Apical four-chamber view demonstrating congenitally corrected transposition of the great arteries. RV, right ventricle; LV, left ventricle; PA, pulmonary artery; B: Modified apical transthoracic echocardiography view. RV, right ventricle; LV, left ventricle; TV, tricuspid valve; MV, mitral valve ; RA, right atrium ; LA, left atrium

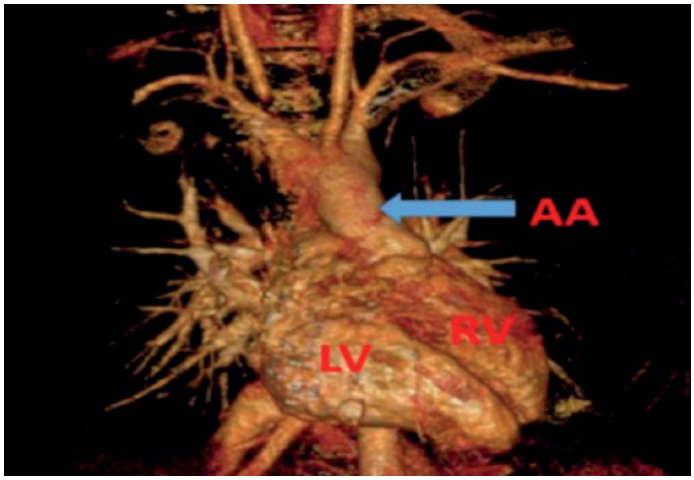

Figure 3. Three-dimensional reconstruct of patient's congenitally corrected transposition of the great arteries anatomy by high resolution computed tomography with anterior view. AA , ascending aorta; RV, right ventricle; LV , left ventricle

DISCUSSION

Congenitally corrected transposition of the great arteries (CCTGA) includes both atrioventricular and ventriculoarterial discordance. Thus, systemic venous blood is carried from the right atrium to the left ventricle and then to the pulmonary artery. Oxygenated blood in the pulmonary veins passes from the left atrium to the right ventricle and then to the aorta. It may be accompanied by VSD, pulmonary stenosis and Ebstein anomaly (1).

Physical examination and echocardiographic examination are very critical in the diagnosis of CCTGA. In physical examination, murmurs due to concomitant cardiac pathologies can be heard. If there is pulmonary stenosis and VSD accompanying CCTGA, cyanosis may occur. The most important aid in diagnosis is the ventriculoarterial discordance along with ventricular inversion in transthoracic echocardiography (1). Magnetic resonance imaging and cardiac CT can be used in the differential diagnosis. In our case, we used cardiac CT to demonstrate the relationship of the great arteries with ventricles.

The prognosis depends on concomitant cardiac defects, systemic ventricular dysfunction and conduction system anomalies. Rhythm disturbances such as severe AV blocks, supraventricular tachycardia, sick sinus syndrome, atrial fibrillation, Wolff-Parkinson-White Syndrome, and ventricular tachycardia can often co-exist (4). In the rhythm holter examination of our case, no anomalies of the conduction system were observed.

In long-term follow-up of these patients, decreased systemic ventricular (right ventricle) functions have been shown to increase mortality. Increased pressure load of the right ventricle, systemic AV valve regurgitation and myocardial hypoxia observed in most patients cause systemic right ventricular dysfunction (2). In our case, there was no defect or conduction disorder accompanying CCTGA. However, there was mild dysfunction in the systemic ventricle.

Anatomical repair of CCTGA prevents systemic RV failure. However, first of all, this strategy can be applied to babies and children until adolescence at the latest, and it has risks and limitations. Also, new challenges may arise in the late postoperative period. Adult patients with CCTGA with advanced systemic RV dysfunction represent the greatest challenge. Various palliative options such as cardiac resynchronization therapy, tricuspid valve repair or replacement, pulmonary artery banding, and assist device implantation to systemic RV can be used to improve functional status and delay the progression of ventricular dysfunction in patients who are not eligible for anatomical correction (5). In symptomatic patients with severe TR and preserved or mildly impaired systemic RV systolic function (EF >40%), TV replacement is indicated. (6). In cases with tricuspid valve regurgitation, tricuspid valve replacement is required before right ventricular dysfunction develops. Studies have suggested that tricuspid valve surgery can provide long-term satisfactory results in patients with CCTGA. In patients with tricuspid valve plastic surgery, recurrent tricuspid regurgitation was observed. Again, it was emphasized that the right end ventricular end-diastolic diameter is a risk factor for late mortality, and for good results, surgery should be performed before systemic ventricular dilatation and dysfunction (7). After right ventricular dysfunction develops in these patients, surgical treatment results are not successful and mortality

is increased. For our patient, due to the mild impairment of the systemic ventricle, the absence of additional defects, and the high risk of anatomical repair, medical treatment was decided. Our patient was started on ACE inhibitor treatment.

In patients with CCTGA, ACE inhibitors and diuretics (spironolactone, furosemide) can be used typically to reduce preload and afterload. Administration of beta-blockers (carvedilol, atenolol) is also beneficial, but they may increase the risk of atrioventricular block as a result of conduction delay. Cardiac glycosides (digoxin) can be used to support systemic RV contractility. Attention should be paid to the risk of bradyarrhythmias and atrioventricular block (5).

In our case the patient is female and most women with ccTGA reach childbearing age. Pregnancy caused changes in the cardiovascular system. For example blood volume increases by 50%, cardiac output increases, cardiovascular resistance reduces. All these changes may lead to right ventricular failure in women with a systemic RV (8). Successful pregnancy can be achieved by most women with ccTGA. However risk factors for pregnancy are systemic RV dysfunction, tricuspid valve regurgitation. Also, complete atrioventricular block is not rare, and those could make worse cardiac function during pregnancy. Nevertheless pregnancy and delivery are well tolerated if RV function is preserved before pregnancy (9). Angiotensin converting enzyme (ACE) inhibitors are contraindicated for women throughout pregnancy because they may have teratogenicity for the fetus (10). On the other hand, because of recurrence risk of the CCTGA in the new born babies, fetal echocardiography or conventional echocardiography after birth should be performed.

CONCLUSION

Isolated CCTGA cases can be asymptomatic for years. However, in patients presenting with dyspnea on exertion, albeit rarely, CCTGA should be kept in mind and should be considered in the differential diagnosis. These patients should be carefully monitored for systemic ventricular and systemic AV valve dysfunction and arrhythmias.

Conflict of interest: The authors declare that they have no competing interest.

Financial Disclosure: There are no financial supports.

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