A Rare Combination of Congenitally Corrected Transposition of the Great Arteries with Pulmonary Artery Aneurysm

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INTRODUCTION

Congenitally corrected transposition of the great arteries (ccTGA) is a rare condition (<1% of all congenital heart disease), and the diagnosis is often unnoticed until adulthood.[1,2] A variety of congenital heart anomalies can also be present with ccTGA. We present the case of a previously undiagnosed patient who developed heart failure symptoms. He was found to have ccTGA accompanied by a large pulmonary artery (PA) aneurysm and an atrial septal defect (ASD), a combination that is rarely seen.

CASE PRESENTATION

A 68-year-old man presented to the emergency department because of worsening dyspnea over the past few months. He reported no medical history. The patient was acyanotic. Physical examination revealed a loud S2, and a holosystolic murmur was heard at the apex. No S3, no rales, and no peripheral edema of the lower limbs were present. Heart rhythm was irregular, and electrocardiography showed atrial fibrillation with intraventricular conduction delay.

Subsequent echocardiographic evaluation revealed ccTGA with situs solitus and mesocardia (Figure 1). The great vessels were side by side (Figure 2), with the aorta lying anteriorly and on the left. The PA featured a large aneurysm up to 6.9 cm, as well as enlarged branches (Figure 3). Two-dimensional echocardiography depicted a right ventricle (subaortic ventricle) supporting the systemic circulation with a tricuspid systemic atrioventricular valve (SAVV). The systemic ventricle (SV) was dilated and hypertrophied, with mildly to moderately impaired ejection fraction (Video 1). Color Doppler echocardiography also detected significant regurgitation of the SAVV, without an Ebstein-like anomaly (Figure 4, Video 2), and a secundum-type ASD with a left-to-right shunt (Figure 5, Video 3). The aortic valve was trileaflet, and there was moderate aortic insufficiency (Figure 4, Video 4). The subpulmonic ventricle (SPV) was a left ventricle connected to the PA, with globally mildly depressed ejection fraction. The pulmonary atrioventricular valve had moderate regurgitation, and the pulmonic valve had mild-to-moderate regurgitation without valvular stenosis (Figure 4, Video 4). The inferior vena cava was connected to the right atrium and was dilated (2.3 cm), with <50% respiratory fluctuation, indicative of high right atrial pressure (estimated mean right atrial pressure 15–20 mm Hg). The pulmonary atrioventricular valve regurgitation jet was insufficient for accurate evaluation of SPV systolic pressure. Pulmonic ventricular diastolic pressure, estimated by Doppler echocardiography, was high. Using the pulmonic valve regurgitation jet end-diastolic velocity (averaging six beats), we calculated SPV diastolic pressure of 35–40 mm Hg. The estimated mean SPV pressure was estimated at 59–64 mm Hg. The aortic root and the proximal ascending aorta were mildly dilated, and both atria were enlarged. A mild-to-moderate pericardial effusion was noted, mainly along the lateral wall of the subaortic ventricle.

The patient was treated medically with diuretics, angiotensin-converting enzyme inhibitors, a novel oral anticoagulant, and a β-blocker and was later referred for cardiac magnetic resonance imaging (CMR), which confirmed the aforementioned findings. CMR depicted a parallel course of the great arteries, with the aorta lying anteriorly and on the left (Figure 3), with a side-by-side orientation of the SV and SPV (Figure 2). The SV was dilated, with an end-diastolic volume of 227 mL (118 mL/m^2) and an ejection fraction of 43%. The SPV had an end-diastolic volume of 159 mL (83 mL/m^2) and an ejection fraction of 45%. Using the aortic and pulmonic stroke volumes by phase-contrast CMR, the Q_p/Q_s ratio was 1.3. There was no persistent left superior vena cava. Regurgitation of both atrioventricular valves was found to be mild to moderate. Contrast-enhanced CMR with myocardial late gadolinium enhancement did not show areas suggestive of replacement fibrosis.

The patient showed improvement on follow-up, and medication doses were titrated accordingly. We recommended imaging the PA aneurysm with cardiac computed tomography in 3–6 months. He was referred to an adult congenital heart disease expert for further evaluation.

DISCUSSION

ccTGA is a congenital anomaly typically characterized by discordant atrioventricular and ventriculoarterial connections and may often remain undiagnosed until adulthood.[3] Associated abnormalities are common, but most frequent among them are pulmonary stenosis (valvular or subvalvular) (40%–74%), ventricular septal defect (70%), systemic atrioventricular (tricuspid) valve abnormalities (90%), and complete heart block (2% per year).[3,4] Only 1% of patients with uncomplicated ccTGA with no recognizable associated anomalies can be diagnosed in late adulthood.[5]

We report the case of a 68-year-old man with ccTGA with a large PA aneurysm and an ASD who presented with heart failure symptoms. Our patient did not develop heart failure symptoms at an

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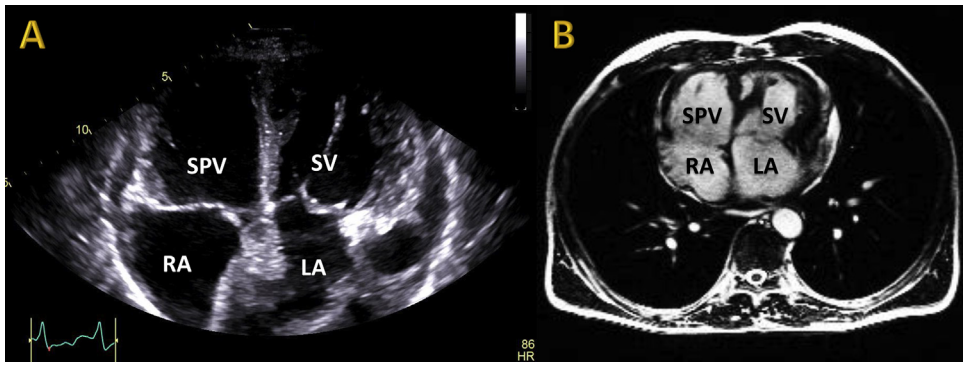

Figure 1 **(A)** Subcostal echocardiographic view with the probe indicator notch to the patient's left. **(B)** CMR showing the cardiac apex pointing to the middle line, depicting situs solitus with mesocardia. *LA*, Left atrium; *RA*, right atrium; *SPV*, subpulmonic ventricle; *SV*, systemic ventricle.

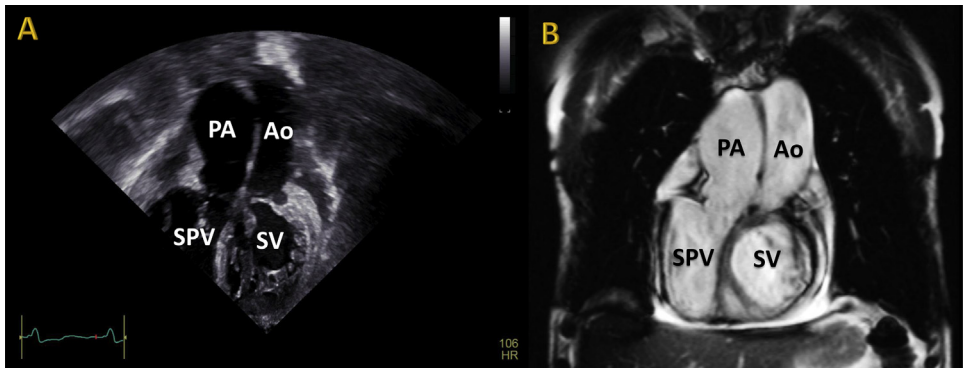

Figure 2 **(A)** Echocardiographic illustration of congenitally corrected transposition of the great arteries. Apex down view and anterior sweep of an apical four-chamber view showing the systemic and subpulmonic ventricle side by side and the great arteries parallel to each other. **(B)** CMR showing the anatomic features of the defect. The systemic ventricle is a right ventricle connected to the aorta and the subpulmonic ventricle is a left ventricle supporting the pulmonic circulation. *Ao*, Aorta; *PA*, pulmonary artery; *SPV*, subpulmonic ventricle; *SV*, systemic ventricle.

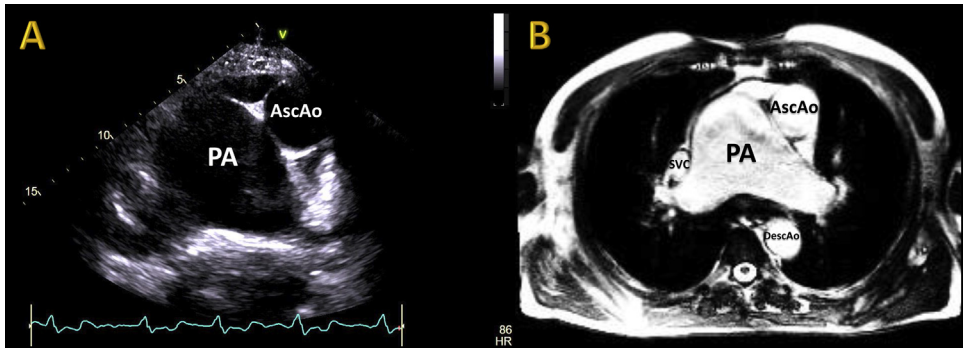

Figure 3 **(A)** An echo short-axis view showing the aorta lying anteriorly and on the left of the pulmonary artery. **(B)** The pulmonary artery aneurysm was measured 6.9 cm by CMR just above the bifurcation of the two dilated pulmonary artery branches. *AscAo*, Ascending aorta; *DescAo*, descending aorta; *PA*, pulmonary artery; *SVC*, superior vena cava.

earlier age, although he had SV dysfunction, SAVV regurgitation, and atrial fibrillation, all of which contribute to a poor outcome.[3] Two thirds of patients with significant coexisting defects have congestive heart failure by 45 years of age.[6] Heart failure syndrome usually develops because the morphologically systemic right ventricle fails to serve as the SV over time.[3] The concomitant SAVV regurgitation is strongly associated with congestive heart failure in these patients, although it is debatable whether its role is primary or secondary.[6] Survival with ccTGA and no coexisting anomalies has been reported to the seventh and eighth decades of life. The usual outcome,

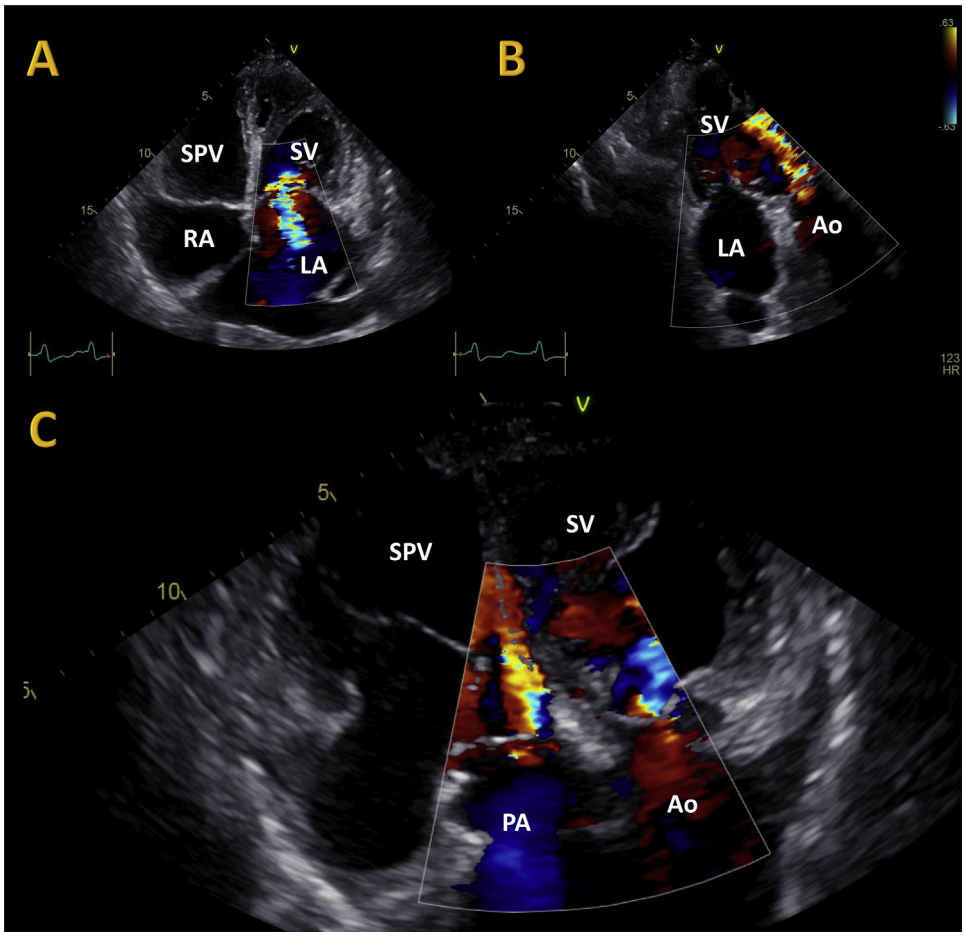

Figure 4 Transthoracic Color Doppler echocardiography. **(A)** Apical four-chamber view showing the significant systemic atrioventricular valve regurgitation. **(B)** Apical three-chamber view showing the moderate aortic valve regurgitation. **(C)** Anterior sweep of an apical four-chamber view illustrating mild-to-moderate pulmonic insufficiency. *Ao*, Aorta; *LA*, Left atrium; *PA*, pulmonary artery; *RA*, right atrium; *SPV*, subpulmonic ventricle; *SV*, systemic ventricle.

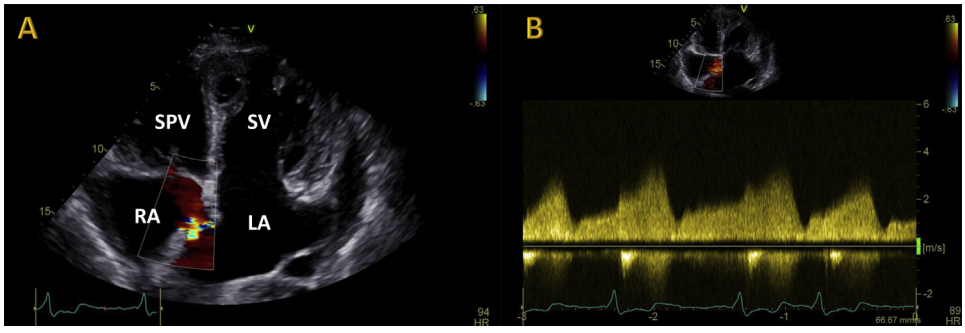

Figure 5 **(A)** Left-to-right shunt via the secundum atrial septal defect is visualized with color Doppler in the apical four-chamber view. **(B)** Continuous wave Doppler, interrogation of flow across the atrial septal defect. *LA*, Left atrium; *RA*, right atrium; *SPV*, subpulmonic ventricle; *SV*, systemic ventricle.

however, is failure of the subaortic ventricle earlier in life, mainly with severe SAVV regurgitation.[3] Many patients present with heart block, and the implantation of a pacemaker is thus very common in ccTGA.[2,4,7] Late complications are usual in patients with ccTGA whether or not they underwent surgical repair, and careful follow-up is necessary.[8,9]

We believe our patient to be a rarity, presenting with ccTGA, a large PA aneurysm, and an ASD. The presence of PA aneurysm with ccTGA has rarely been described before,[10] and their combination with an ASD has, to our knowledge, been previously reported only once, in a patient with dextrocardia.[11] Isolated PA aneurysms are very rare, but they are more common when combined with congenital heart

defects.[12] Most aneurysms are diagnosed incidentally on chest radiography or computed tomography.[13] Their diagnosis is important because of high mortality upon dissection or rupture.[14] The long-standing ASD, in addition to elevated SV pressure and increased left atrial pressure, likely led to elevated PA pressures in our patient, and this partly added to the pathology of the PA aneurysm. Increased afterload of the SPV in patients with ccTGA may reduce septal shift, reduce SAVV regurgitation grade, and alleviate heart failure symptoms.[15]

CMR confirmed the diagnosis and provided an accurate anatomic and functional evaluation of this complex congenital defect. CMR is a useful modality in complex congenital heart disease for accurate assessment of SV failure, atrioventricular valve regurgitation grading severity, and the presence of myocardial replacement fibrosis.

Cardiac catheterization may be required, especially in older patients, to detect significant coronary artery disease and assess the coronary anatomy for associated anomalies (single coronary artery). Left and right heart hemodynamics can be obtained to accurately determine pulmonary and systemic artery pressures, intracardiac pressures, mean pulmonary capillary wedge pressure, and oxygen saturation at different levels and to calculate the shunt across defects. The recorded hemodynamic data may be critical to decide upon the therapeutic approach. There is no clear recommendation for the best treatment strategy in patients with ccTGA and associated abnormalities. Although it was not unreasonable to catheterize our patient, we suggested reevaluation with echocardiography and reassessment of PA aneurysm size with cardiac computed tomography in 3–6 months, as he markedly improved with medical treatment, and no compression of adjacent structures or thrombus formation in the PA was noted on CMR.

CONCLUSION

We present the case of a patient with ccTGA that was diagnosed at an advanced age, despite the complexity of the anatomic defects. This combination of ccTGA with a PA aneurysm and an ASD is considered extremely rare.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at https://doi.org/10.1016/j.case.2018.04.006.

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