

A case report of late physiologic repair of congenitally corrected transposition of the great arteries and pulmonary stenosis in a severely cyanotic patient: better late than never

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Background

Patients with congenitally corrected transposition of great arteries (ccTGA) not infrequently seek medical attention for the first time late in life. Optimal management of natural history ccTGA is debated and must be tailored.

Case summary

A 38-year-old male patient was referred to our centre because of severe cyanosis and worsening dyspnoea. Investigations disclosed situs solitus, mesocardia, double discordance, large ventricular septal defect (VSD), severe pulmonary stenosis, and no significant atrio-ventricular valves regurgitation. The patient underwent physiologic repair: VSD closure, placement of a left ventricle to pulmonary artery conduit, and epicardial atrio-biventricular pacemaker implantation. The conduit was intentionally undersized to promote tricuspid valve continence. Post-operative course was uneventful, transthoracic echocardiography showed good biventricular function without significant tricuspid regurgitation. At 1 month after discharge, the patient is in New York Heart Association Class II.

Discussion

Management of late presenter patients with ccTGA depends on the associated lesion and estimation of surgical risk. In selected patients markedly symptomatic physiologic repair is a rationale option, providing a normal saturation and biventricular circulation with a significantly lower surgical risk as compared with an anatomic repair.

Keywords

Congenitally corrected transposition of the great arteries • Adult congenital heart disease • Cyanosis • Pulmonary stenosis • Ventricular septal defect • Surgery • Physiologic repair • Case report

ESC Curriculum

2.1 Imaging modalities • 9.7 Adult congenital heart disease • 7.5 Cardiac surgery

Learning points

- Heart failure and severe cyanosis is a common presentation of adult patients with unrepaired congenitally corrected transposition of great arteries, ventricular septal defect, and severe pulmonary stenosis surviving to adulthood.
- Complete repair and achievement of biventricular circulation is feasible and should be the preferred option even at the late stage of the disease.
- Physiologic repair, leaving a morphologic right ventricle as systemic pumping chamber, may be preferred over anatomic repair in selected cases.

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Introduction

Not infrequently, patients with unrepaired complex congenital heart diseases are referred to adult congenital heart disease (ACHD) centres for the first time because of arrhythmias, heart failure, or complication of severe cyanosis.^{1,2}

The decisional process in this clinical setting is particularly challenging, as the physiology and mechanisms of adaptation unicity prevent adherence to a standardized guideline. In patients with anatomies within the spectrum of congenitally corrected transposition (ccTGA) the associated lesions, i.e., ventricular septal defect (VSD) and pulmonary stenosis (PS) dictate treatment strategy. Whenever the clinical picture is dominated by severe cyanosis, additional risks due to coagulation cascade abnormalities and chronic polyglobulia must be taken into account. Conservative medical management or palliative procedures, aiming at increasing pulmonary blood flow, are usually preferred because of the feeling of a prohibitive risk portended by complete repair.³

We herein report a case of successful physiologic repair of a ccTGA, VSD, and severe pulmonary stenosis that allowed restoration of a biventricular circulation and normal blood saturation.

Timeline

Day 1	Referred for severe cyanosis and worsening effort dyspnoea
Day 2	Cardiac magnetic resonance disclosed double discordance ventricular septal defect (VSD), severe pulmonary stenosis, balanced well-functioning right and left ventricles
Day 4	Diagnostic catheterization revealed low pulmonary and systemic ventricle end-diastolic pressure
Day 6	Physiologic repair was performed: VSD closure aligning aortic valve with left-sided morphologic right ventricle. Left ventricle to pulmonary artery conduit was used to create the sub-pulmonary outflow tract and an atrio-biventricular pacemaker was implanted
Days 7–9	Intensive care unit stay. Extubated on the second post-operative day. Weaned from low dose adrenaline support.
Day 20	Echocardiography showed good biventricular function and only mild tricuspid regurgitation. There was no evidence of congestion on clinical examination. The patient was discharged on furosemide 50 mg daily.
Day 50	Follow-up echocardiography showed persistent good biventricular function. Although, the patient remained mildly breathless with New York Heart Association Class II symptoms

Case presentation

A 38-year-old Tunisian man was admitted because of severe cyanosis and effort intolerance.

At the age of 2 years, a complex cyanotic cardiac defect was diagnosed and left un-operated. He was lost at follow-up, thereafter. Due to worsening cyanosis and minimal effort dyspnoea, he was admitted to a local hospital and referred, thereafter, to our centre.

At admission, the patient was deeply cyanotic (O₂ Saturation at finger plethysmography was 72%), with clubbing. On physical examination a ubiquitous systolic 3/6 murmur was appreciated, lungs were clear, and no peripheral congestion was observed. Electrocardiogram (ECG) showed complete heart block, atrio-ventricular (AV) dissociation with junctional escape rhythm (heart rate 60/min) increased QRS voltages and deep S waves in the precordial leads (Figure 1). Haematocrit was 65% with iron storage within the normal range.

The echocardiographic anatomic diagnosis was: situs solitus, mesocardia, double discordance (i.e. AV and ventriculoarterial discordance) with, large VSD, and severe pulmonary stenosis with biventricular hypertrophy and good systolic function (Videos 1–3, Supplementary material online, Video S1).

Cardiac computed tomography confirmed atrial, ventricular, and great vessels arrangement and normal pulmonary drainage (Figure 2). Cardiac magnetic resonance showed balanced well-functioning morphological right and left ventricles, no significant AV valves regurgitation, normal pulmonary vasculature, and occlusion of the right brachiocephalic vein at the junction with the superior vena cava (SVC), with venous flow collateralization into distal SVC through the azygos system (Figures 3 and 4, Supplementary material online, Video S4).

Cardiac catheterization demonstrated low pulmonary and systemic ventricle end-diastolic pressures (mean pulmonary artery pressure 18 mmHg), unobstructed epicardial left- and right-coronary arteries, correctly originating from the respective facing sinuses and absence of significant collateral circulation. Pulmonary angiogram confirmed pinpoint pulmonary stenosis and post-stenotic pulmonary artery dilatation (Figure 5).

Owing to the extreme symptomatic hypoxia, a physiologic repair contemplating VSD closure, relief of pulmonary stenosis, and pacemaker implantation was planned. This option was discussed with the patient that gave his consent.

Surgery was carried out by median sternotomy. Cardiopulmonary bypass (CBP) was realized by bicaval and aortic cannulation. Ventricular septal defect was closed through the right-sided mitral valve, aligning the left-sided morphological right ventricle (RV) with the aorta. The pulmonary outflow was reconstructed by the interposition of an 18 mm valved conduit (Hancock® 18) between the right-sided morphological left ventricle (LV) and the pulmonary artery (PA). The conduit was intentionally undersized in order to realize a moderate banding of the sub-pulmonary ventricle aiming to promote tricuspid continence. A 4 mm calibrated interatrial communication was left. Finally, an epicardial atrio-biventricular pacing system was implanted in order to avoid pacing-related mechanical desynchronization.

Separation from bypass was easily achieved with a low dose of adrenaline (0.04 mcg/Kg/min). Cardiopulmonary bypass and aortic cross-clamp times were 90' and 53' min, respectively.

The patient was weaned from the ventilator and extubated on the second post-operative day. The postoperative course was uneventful. Transthoracic echo revealed good biventricular function, mild

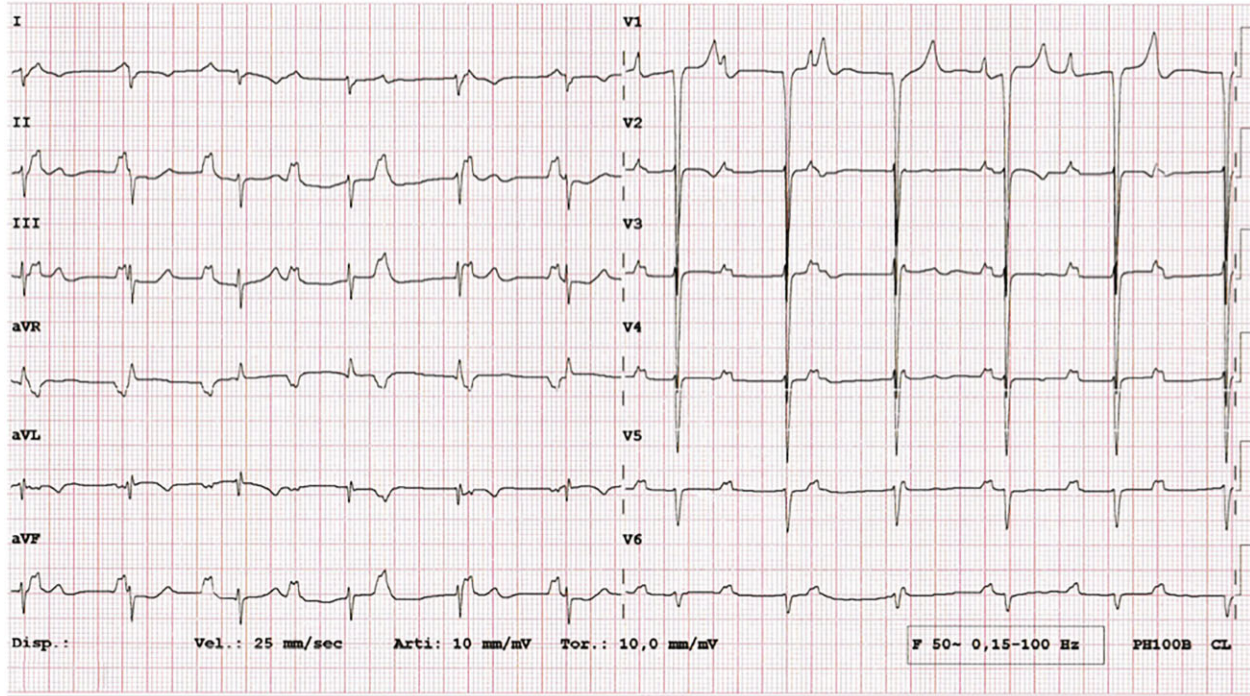


Figure 1 Twelve leads electrocardiogram recording. Atrio-ventricular dissociation with junctional escape rhythm is shown.



Video 1 Subcostal four-chamber 2D-colour view showing hypertrophied well-functioning left-sided morphological right ventricle and right-sided morphological left ventricle. Colour Doppler demonstrates bidirectional flow across a wide non-restrictive ventricular septal defect.

tricuspid regurgitation, and mean gradient across the LV-PA conduit was 30 mmHg. (Supplementary material online, Videos S2 and S3) Initial inferior vena cava congestion with evidence of dense spontaneous echo-contrast, progressively resolved along with haematocrit normalization and venous pressure reduction.

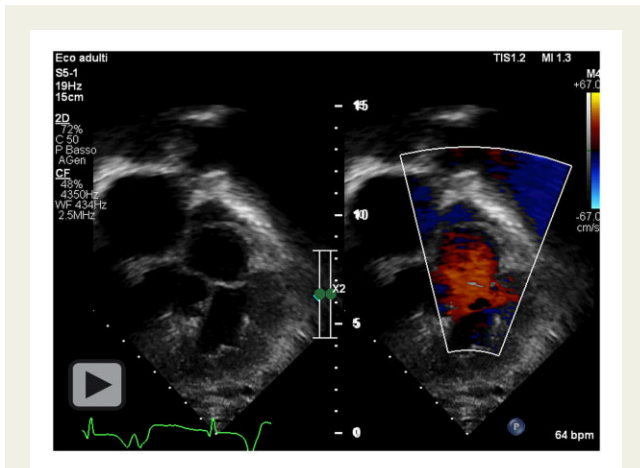
At discharge, on 20th post-operative day, the patient was asymptomatic without evidence of congestion on 25 mg of Furosemide

b.i.d. and 25 mg of Spironolactone o.d., saturation was constantly above 95% even during moderate efforts. Close clinical and echocardiographic follow-up was arranged locally once the patient was back home, in order to monitor congestion and titrate diuretic therapy. At last contact with the local cardiology team, 50 days after the operation, ventricular function was stable, and the patient was well compensated on 40 mg of Furosemide.

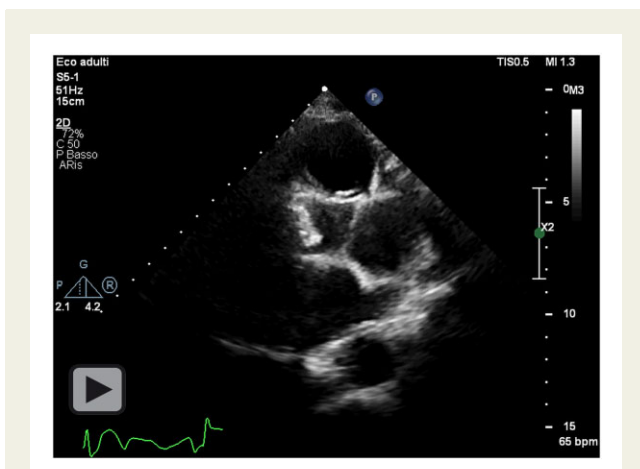
Discussion

Congenital corrected transposition of great arteries is a rare disease that not infrequently is diagnosed for the first time during adulthood.⁴

Management depends mainly on the associated lesions. When the patient presents with cyanosis due to severe pulmonary stenosis theoretical therapeutic options are anatomic repair, by performing an atrial switch combined with a Rastelli procedure, or physiologic repair, by closing the VSD, relieving sub-pulmonary obstruction, leaving the left-sided morphologic RV as systemic pumping chamber. Uni-ventricular palliation has been recommended whenever anatomical constraints, such as major valvular straddling or ventricular hypoplasia prevents biventricular repair.⁴ It should be pointed out that the wording 'physiologic repair' in this context might be misleading. Indeed, this term is conventionally used to indicate an arrangement where systemic and pulmonary circulations are in series and separated although, differently from anatomic repair, the morphologic RV became the systemic pumping chamber, which is, by definition, a non-physiologic condition.



Video 2 Sub-costal four-chamber 2D-colour view showing continent tricuspid valve.



Video 3 Short axis illustrating great arteries alignment, with aortic valve anterior and to the left of the fibrotic and thickened pulmonary valve.

Although more technically challenging, anatomic repair has been advocated by some institutions as a first choice approach based on data suggesting an excess of adverse events in patients who had undergone physiologic correction.⁵⁻⁷

The main issue against this latter option is the concern of late morphologic RV failure and tricuspid regurgitation. On the other hand, both double switch and atrial switch-Rastelli procedure are burdened by a not negligible morbidity and mortality, particularly in late presenter patients.⁸

We elected our patient to undergo physiologic repair based on three arguments. Firstly, we assumed as main clinical target of the intervention the improvement of saturation throughout the most definitive and straightforward operation, taking into consideration the patient age and the long-standing severe desaturation, that would have increased the post-operative risk of bleeding and target organ failure. In the second instance, the patient presented with a

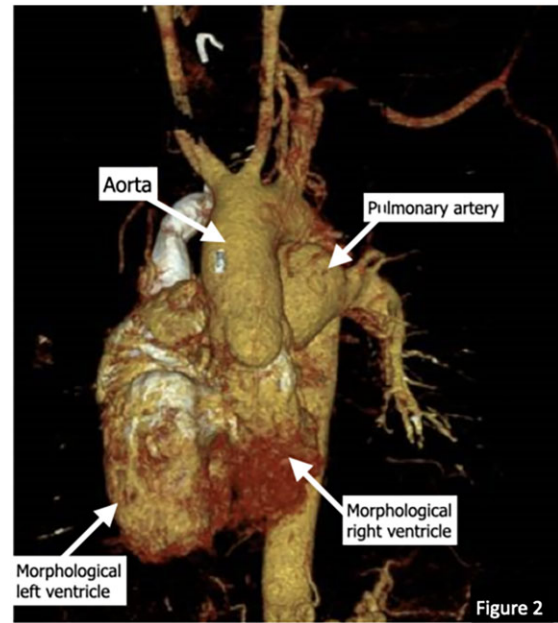


Figure 2 Computed tomography 3D reconstruction of the anatomy showing ventricular mass and malposition of the great arteries.

hypertrophic well-functioning morphologic RV without significant tricuspid regurgitation, encouraging physiologic repair. Furthermore, we presumed that leaving moderate sub-pulmonary obstruction might have granted tricuspid competence, by realigning the inter-ventricular septum and avoiding tricuspid septal leaflet displacement. In a previous report, an adult patient with a similar anatomic arrangement, necessarily underwent anatomic repair because of severe systemic (tricuspid) AV valve regurgitation.⁹ However, in the aforementioned case, the clinical picture was dominated by heart failure and congestion due to systemic ventricle dilatation and tricuspid regurgitation. In our patient, the key clinical and anatomic features were severe cyanosis, caused by severe pulmonary stenosis with hypertrophic not dilated ventricles. According to this pathophysiologic background, we felt that the creation of a left ventricle to aorta pathway would have left a relatively hypoplastic and restrictive RV.

There is very limited experience of late physiologic repair of ccTGA.¹⁰ This case presents a combination of unique features in terms of patient age, degree of cyanosis and symptoms severity, illustrating how patient-specific characteristics should guide a tailored treatment, targeting the dominant mechanism of disease. In particular, from the pathophysiologic point of view, the combination of large VSD and severe PS allowed both ventricles to properly develop, granting a balanced circulation and preventing the occurrence of pulmonary vascular disease.

Physiologic repair of ccTGA with VSD and severe PS is a feasible and valid alternative to anatomic repair, achievable with a significantly shorter CBP and cross-clamping time, in selected patients deemed to be at high operative risk, symptomatic for long-standing profound desaturation, with balanced well-functioning ventricles and continent systemic AV valve.

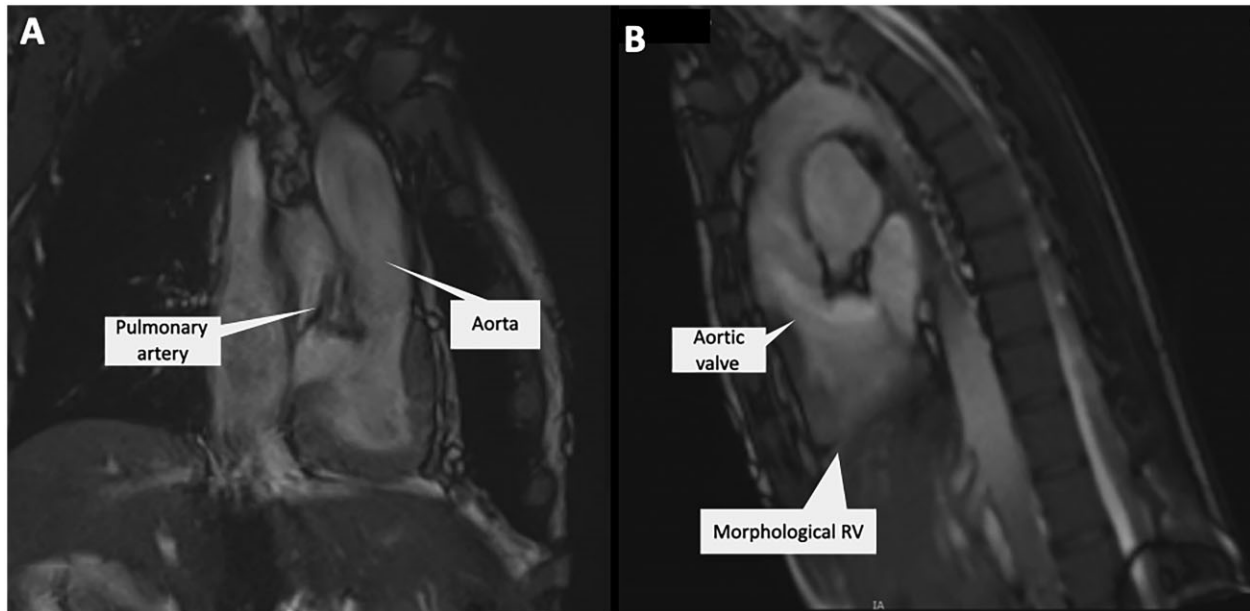


Figure 3 (A) Cardiac magnetic resonance cine showing both stenotic sub-pulmonary and systemic outflows. (B) Cardiac magnetic resonance cine long-axis view showing anterior aorta arising from morphological right ventricle.

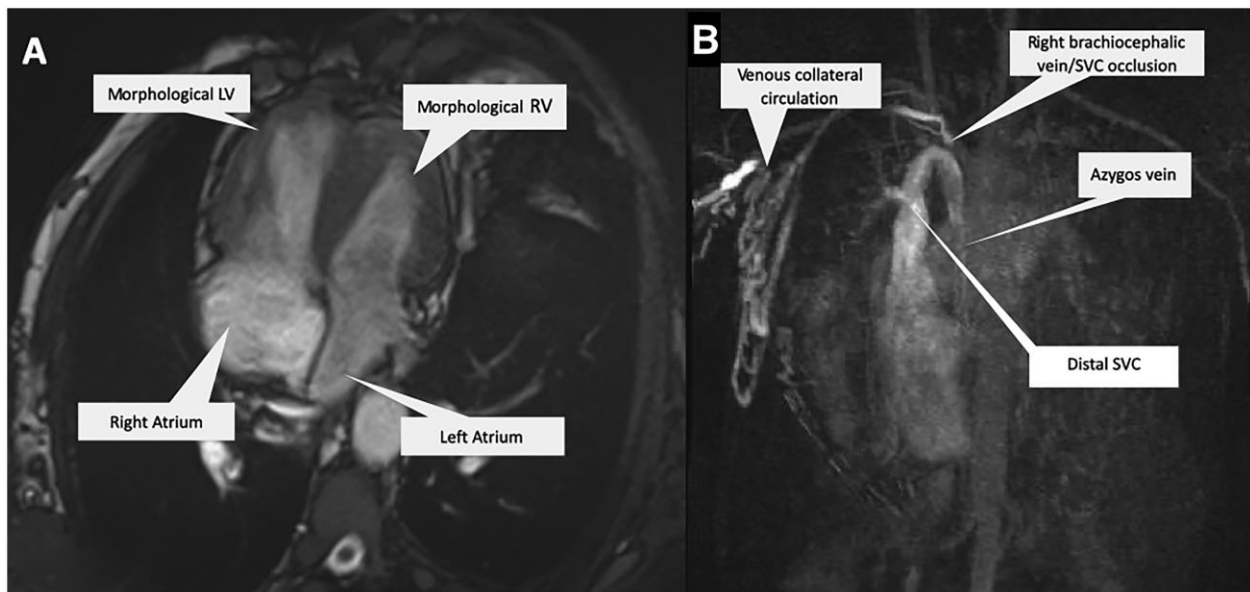


Figure 4 (A) Cardiac magnetic resonance cine four-chamber view showing well-formed and balanced atria and ventricles. (B) Twisted sequence demonstrating occlusion of right brachiocephalic vein at the junction with the superior vena cava and collateralization into the distal superior vena cava throughout the azygos system.

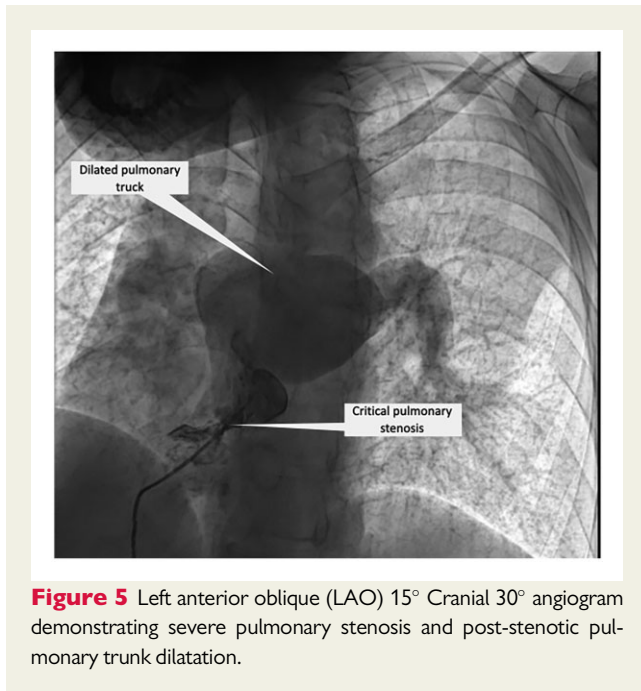


Figure 5 Left anterior oblique (LAO) 15° Cranial 30° angiogram demonstrating severe pulmonary stenosis and post-stenotic pulmonary trunk dilatation.

Finally, it should be appreciated that the late repair of adults with complex congenital heart disease should contemplate the arrangement of close follow-up as the risk of developing heart failure remains substantial.

Lead author biography



Dr Paolo Ferrero is a pediatric and adult congenital heart disease cardiologist. I am currently working in the pediatric and adult congenital heart disease unit at IRCCS, S. Donato, Milan. My main areas of interest are diagnosis and treatment of congenital heart disease, with a focus on pulmonary hypertension, advanced heart failure, and arrhythmias. I have also an interest in prognostic modelling and evidence appraisal in congenital heart disease.

Supplementary material

Supplementary material is available at *European Heart Journal—Case Reports* online.

Slide sets: A fully edited slide set detailing these cases and suitable for local presentation is available online as [Supplementary data](#).

Consent: The authors confirm that written consent for the submission and publication of this case, including images, has been obtained from the patient in line with COPE guidance.

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