CASE REPORT

An adult with a sinus venosus atrial septal defect and dilated cardiomyopathy

Luke Oakley,¹ Sean Foley,² Justin Cox,¹ Daniel Seidensticker¹

¹Department of Cardiology, Naval Medical Center, San Diego, California, USA
²Department of Internal Medicine, Naval Medical Center, San Diego, California, USA

Correspondence to
 Dr Luke Oakley,
 luke.s.oakley@gmail.com

Accepted 3 April 2014

SUMMARY

Dilated cardiomyopathy, heart failure and atrial septal defects are well-recognised entities in isolation, but are rarely seen together. Now that 90% of children with congenital heart disease survive into adulthood, such combinations of disease are increasingly seen in adult cardiology. While most young patients with dilated cardiomyopathy respond well to medical therapy, some do not, and require more invasive management. We describe a 32 year-old man with dilated cardiomyopathy and a sinus venosus-type atrial septal defect associated with a remarkable pulmonary to systemic flow ratio of 5:1. We propose that the atrial septal defect blunted his heart failure symptoms by serving as a 'pop-off' valve and limiting pulmonary congestion. The patient ultimately failed medical management and received a left ventricular assist device. The case is presented along with a discussion of this unique pathophysiology and a brief review of the literature in this rapidly evolving field.

BACKGROUND

Owing to the therapeutic improvements in congenital heart disease it is now estimated that 90% of these patients survive into adulthood, and that the number of adults with congenital heart disease now outnumbers that of children. As a result, congenital defects and their combinations represent an emerging trend in adult cardiology.¹

This case illustrates the complexity of treatment modalities for adult patients with congenital heart malformations, and seeks to better characterise a novel mechanism of how these defects can influence other derangements of cardiac physiology.

CASE PRESENTATION

A 32-year-old man presented with 2 weeks of progressive dyspnoea on exertion, 10 kg weight gain and paroxysmal nocturnal dyspnoea. He sought medical care 1 week prior for a presumed upper respiratory infection, and was treated unsuccessfully with inhaled albuterol, azithromycin and corticosteroids. One month prior he was able to run 1.5 miles without difficulty. Medical history was only notable for smoking six cigarettes per day for 8 years, and binge drinking up to 10 drinks, 2–3 times per month, although he had cut back considerably in the preceding year. He denied use of nutritional supplements, over-the-counter medications and illicit drugs. Family history was unremarkable.

His blood pressure was 108/72 mm Hg, heart rate was 118 bpm and respirations were laboured at 32 breaths per minute. Inspection of the chest was

significant for a protuberance of the left parasternal chest wall, and cardiac auscultation revealed a widely split and fixed S2. The pulmonary examination was notable for bibasilar rales and dullness to percussion in the lower lung fields. There was 9 cm of jugular venous distension and 5 mm of pitting oedema up to the mid-calf bilaterally. These signs and symptoms were consistent with right and left heart failure.

INVESTIGATIONS

Laboratory studies were obtained and the values are shown in the table below:

Test	Result value	Reference range
NT-proBNP	6280 pg/mL	<100
ALT	141 units/L	13–69
Troponin-I	0.028 µg/L	<0.034
Urine drug screen	Negative	N/A

ECG revealed premature ventricular contractions, poor R-wave progression, right bundle branch block and a QTc of 457 ms. Chest X-ray ([figure 1](#)) demonstrated only cardiomegaly. Transthoracic echocardiogram revealed severe left ventricular enlargement, dilated cardiomyopathy (DCM) and an aneurismal atrial septum with an atrial septal defect (ASD; [figure 2](#)). The ejection fraction (EF) was visually estimated as 5–10% on the initial ECG, and was confirmed as 5.6% on the cardiac MRI. Cardiac-gated CT

Figure 1 Chest X-ray. Posteroanterior view showing cardiomegaly and pulmonary vascular engorgement.

To cite: Oakley L, Foley S, Cox J, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2013-201306

Figure 2 Transthoracic echocardiogram. Apical four-chamber view at end diastole, shows severe left ventricular enlargement and dilated cardiomyopathy.

Figure 3 Cardiac-gated CT. Reformatted and enhanced oblique view showing the escape of contrast from the left atrium (LA) to the right atrium (RA) through the atrial septal defect (arrow).

(figure 3 and video 1) and MRI (figure 4 and video 2) further demonstrated normal coronary arteries, confirmed a sinus venosus ASD and estimated a remarkable pulmonary to systemic flow ratio (Qp:Qs) of 5:1, which was later confirmed by right heart catheterisation. The pulmonary capillary wedge pressure was 20 mm Hg and the mean pulmonary artery pressure was 35 mm Hg. The cardiac MRI failed to show any delayed hyper-enhancement which would indicate ischaemia, myocarditis or an infiltrative process. Neither imaging technique revealed anomalous pulmonary veins, which are frequently associated with sinus venosus defects.

DIFFERENTIAL DIAGNOSIS

Even when a diagnosis of heart failure is carried out, determining the aetiology often remains elusive. While coronary artery

Figure 4 Cardiac MRI. Markedly dilated ventricles (left ventricle shown on bottom in this orientation) with diffuse wall thinning. Global hypokinesia is evident when comparing the systole (A) with the diastole (B). The ejection fraction on this study was calculated as 5.6%.

Video 1 Cardiac gated CT. Reformatted and enhanced oblique view shows the escape of contrast from the left atrium (LA) to the right atrium (RA) through the atrial septal defect (arrow).

disease and hypertension account for approximately 75% of all heart failure cases, the remaining 20–30% results from a wide variety of aetiologies including viral infection, drug and alcohol use, infiltrative and inflammatory diseases and congenital malformations.²

This patient's age, lack of angina and pattern of DCM made coronary artery disease and ischaemic cardiomyopathy unlikely. His rapid progression and lack of preceding high-blood pressure made hypertensive heart disease unlikely. Alcoholic cardiomyopathy was less likely due to minimal intake prior to presentation.

The large ASD and chest wall deformity suggested chronic remodelling from this congenital malformation. In contrast, his rapid decline, symptoms following a respiratory illness and profound DCM with prominent left ventricular dysfunction supported an acute viral or idiopathic cardiomyopathy, and would be largely inconsistent with cardiac remodelling from the ASD.

TREATMENT

The primary therapy for DCM is neurohormonal blockade with ACE inhibitors, β -blockers and aldosterone blockade. An implantable cardioverter defibrillator (ICD) is also recommended for the primary prevention of life-threatening ventricular arrhythmias for patients with New York Heart Association (NYHA) class II to III symptoms and EF $\leq 35\%$ after a trial of optimal medical management. Similarly, biventricular pacing (also known as cardiac resynchronisation therapy) is indicated for patients with an EF $\leq 35\%$, sinus rhythm, left bundle branch block (LBBB) with a QRS duration ≥ 150 ms and NYHA class II or worse symptoms.³ Those with non-LBBB patterns (as in the patient presented here), or shorter QRS complexes can be

Video 2 Cardiac MRI. Markedly dilated ventricles with diffuse wall thinning in a oblique four-chamber view showing the right atrium (RA), right ventricle (RV), left atrium (LA), and left ventricle (LV). Global hypokinesia is evident with an ejection fraction of 5.6%.

considered for cardiac resynchronisation therapy, but face less certain benefits from this therapy. When these measures fail, patients should be referred for cardiac transplantation or placement of a left ventricular assist device (LVAD).

Recent advances in LVAD technology have made them a safe, effective and practical adjunct to traditional therapy and have expanded the possibilities for patients whose recovery was previously limited by the supply of donor hearts. Patients should be considered for implantation of an LVAD when they remain symptomatic with NYHA class III or IV symptoms or an EF <25% despite optimal medical therapy cannot tolerate medical therapy or have frequent rehospitalisations.⁴

LVADs are primarily used in three capacities: a bridge to transplant, a bridge to recovery with eventual explantation of the device after recovery or as 'destination therapy' to improve functional status in patients not suitable for transplant. As a bridge to recovery, they have shown the potential to reverse heart failure by off-loading stress on the left ventricle while pharmacotherapy promotes remodelling.⁵⁻⁶ They have proven long-term survival and quality-of-life benefit.⁷ Based on manufacturer recommendations, oral anticoagulation is recommended for patients with contemporary LVAD models to minimise the risk of thromboembolism.⁸

Most patients with ASDs should undergo closure except in the setting of severe irreversible pulmonary hypertension without left-to-right shunting (Eisenmenger physiology)—when closure would result in acute decompensation.⁹ Closure was shown to be superior to medical therapy in a study of 179 patients over 40 years. Functional improvement was seen in 32% of the 84 patients who underwent closure as compared with only 3% of the 95 who did not. Survival after 10 years was also higher following closure; 95% of patients who underwent closure survived, compared with only 84% of those who did not.¹⁰

OUTCOME AND FOLLOW-UP

Neurohormonal blockade was initiated with an optimal medical regimen, but NYHA class III symptoms persisted. His large stature made matching for a cardiac transplant highly unlikely, so a Heartmate II LVAD and a dual-chamber ICD were implanted and the ASD was closed with a pericardial patch. Intraoperatively, the ASD was measured at 3 cm in diameter.

After these interventions the patient's symptoms rapidly improved and he now enjoys NYHA class I functional status. While his clinical improvement is gratifying, it remains unknown how he will respond to the LVAD with continued use—at the time of writing his EF has improved minimally, necessitating continued use of the LVAD.

DISCUSSION

Heart failure is uncommon in young patients, yet early diagnosis is critical so that appropriate interventions can be made. This case illustrates the challenges of this diagnosis, shows the dramatic consequences of cardiomyopathy in young adults and presents the ASD as a possible relief communication that may have prevented a more fulminant course—a novel pathophysiological mechanism.

While heart failure physiology is well characterised, the role that ASDs play in its progression is not well defined. Although ASDs account for 25–30% of congenital heart defects discovered in adulthood, the sinus venosus subtype is relatively rare, accounting for only 5–10% of these defects.¹¹ In isolation, the symptoms and complications of ASDs largely depend on their size and the degree to which they shunt blood from the systemic (Qs) to pulmonary circulation (Qp): the Qp:Qs ratio. They most often present with atrial arrhythmias, right ventricular failure or paradoxical embolisation.¹

A shunt is considered significant when the Qp:Qs is greater than 1.5:1 or if it causes dilation of the right heart chambers.¹² The Qp:Qs ratio can be calculated based on flow patterns

Figure 5 Pulmonary to systemic flow ratio (Qp:Qs) derivation using two-dimensional echocardiography and spectral Doppler flow measurements. (A) Depicts measurement of the pulmonary flow (Qp) in the parasternal short-axis view at the level of the aortic valve. (B) Depicts measurement of the systemic flow (Qs) in the parasternal long axis. The velocity time integral (VTI) represents velocity over time. It is represented by the spectral flow measurements shown on the right side of parts (A) and (B) of the figure. The area under the curve thus represents the distance travelled by the blood cells. The flow (Q) is calculated by multiplying the outflow tract diameter with this distance (the area under the VTI curve). (Graphics, courtesy of the Canadian Society of Echocardiography.)

measured using a variety of imaging modalities. Figure 5 illustrates the calculation of the Qp:Qs using echocardiographic data. Remarkably, this patient had a Qp:Qs of 5:1. In a report of similar cases, the largest Qp:Qs measured was 4.4.¹³

Here we propose that the ASD may have functioned as a relief communication, a theory proposed elsewhere as a 'pop-off valve', but not previously illustrated with such a case.^{13 14}

We suggest that this patient likely experienced a gradual and asymptomatic process of cardiac remodelling from the congenital ASD, lasting for years until this acute decompensation. When the second hit occurred, a likely viral or idiopathic cardiomyopathy, left atrial pressures increased to aid the failing ventricle leading to greater flow across the ASD. By allowing recirculation of shunted blood in the setting of DCM and severely reduced left ventricular function, the ASD thus may

have limited pulmonary congestion which could have otherwise led to fulminant cardiac failure.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Sommer RJ, Hijazi ZM, Rhodes JF. Pathophysiology of congenital heart disease in the adult, part I: shunt lesions. *Circulation* 2008;117:1090–9.
- 2 Mann DL. Heart failure and cor pulmonale. In: Fauci AS, Longo DL, Eugene K, et al. eds. *Harrison's principles of internal medicine*. 17th edn. New York: McGraw-Hill, 2008.
- 3 Brignole M, Auricchio A, Baron-Esquivias G, et al. 2013 ESC Guidelines on cardiac pacing and cardiac resynchronization therapy. *Eur Heart J* 2013;34:2281–329.
- 4 Lindenfeld J, Albert NM, Boehmer JP, et al. HFSA 2010 comprehensive heart failure practice guideline. *J Card Fail* 2010;16:e1.
- 5 Birks EJ, Tansley PD, Hardy J, et al. Left ventricular assist device and drug therapy for the reversal of heart failure. *N Engl J Med* 2006;355:1873–84.
- 6 Birks EJ, George RS, Firouzi A, et al. Long-term outcomes of patients bridged to recovery versus patients bridged to transplantation. *J Thorac Cardiovasc Surg* 2012;144:190–6.
- 7 Rose EA, Geligns AC, Moskowitz AJ, et al. Long-term use of a left ventricular assist device for end-stage heart failure. *N Engl J Med* 2001;345:1435–43.
- 8 Suarez J, Patel C, Felker M, et al. Mechanisms of bleeding and approach to patients with axial-flow left ventricular assist devices. *Circ Heart Fail* 2011;4:779–84.
- 9 Lindsey JB, Hillis LD. Clinical update: atrial septal defect in adults. *Lancet* 2007;369:1244–6.
- 10 Konstantinides S, Geibel A, Olschewski M, et al. A comparison of surgical and medical therapy for atrial septal defect in adults. *N Engl J Med* 1995;333:469–73.
- 11 Warnes AC, Williams RG, Bashore TM, et al. ACC/AHA 2008 guidelines for the management of adults with congenital heart disease. *Circulation* 2008;118:714–833.
- 12 Webb G, Gatzoulis MA. Atrial septal defects in the adult: recent progress and overview. *Circulation* 2006;114:1645–53.
- 13 Ennezat PV, Juthier F, Marechaux S, et al. Atrial septal defect associated with dilated cardiomyopathy in the setting of acute cardiac failure: importance of comprehensive bedside echocardiography in the ICU. *Eur J Echocardiogr* 2010;11:E11.
- 14 Attenhofer Jost CH, Connolly HM, Danielson GK, et al. Sinus venosus atrial septal defect: long-term postoperative outcome for 115 patients. *Circulation* 2005;112:1953–8.

Learning points

- The incidence of congenital heart disease in adults will continue to rise as more paediatric patients survive into adulthood.
- Cardiologists and primary care physicians must be aware of the potential complications in their respective populations.
- Left ventricular assist devices have shown great success in treating heart failure, and should be considered when medical management fails.
- Adults with congenital heart malformations may present differently than patients with normal hearts, and practitioners should be aware of how these anatomical differences may cloud the diagnosis.
- In some cases, atrial septal defects may serve as a 'pop-off' valve, limiting pulmonary congestion via a left to right shunt, and actually mitigating symptoms.

Copyright 2014 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- Submit as many cases as you like
- Enjoy fast sympathetic peer review and rapid publication of accepted articles
- Access all the published articles
- Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow