A rare case of congenital heart disease with first presentation in adulthood

Introduction

The presence of cardiac symptoms coupled with an abnormal electrocardiogram in the young adult is not an uncommon clinical scenario encountered on the acute medical take or in the general cardiology clinic. The differential diagnosis is vast and this case illustrates the importance of thinking beyond the common causes and considering some of the rarer conditions, such as late presenting adult congenital heart disease.

Discussion

Congenitally corrected transposition of the great arteries represents 0.5% of all cases of congenital heart disease. The condition is characterized by both atrioventricular and ventriculo-arterial discordance. This results in the left atrium connecting to the morphological right ventricle via the tricuspid valve. The right ventricle in turn connects to the aorta supplying the systemic circulation. The right atrium is connected to the morphological left ventricle via the mitral valve. This ventricle supplies the pulmonary circulation via the pulmonary artery.

The condition is 'corrected' as deoxygenated venous blood is supplied to the lungs and oxygenated blood is supplied to the systemic circulation. This phenomenon does not occur often in isolation and is usually found in association with other intracardiac defects, most commonly ventricular septal defects (Hornung and Calder, 2010). In this

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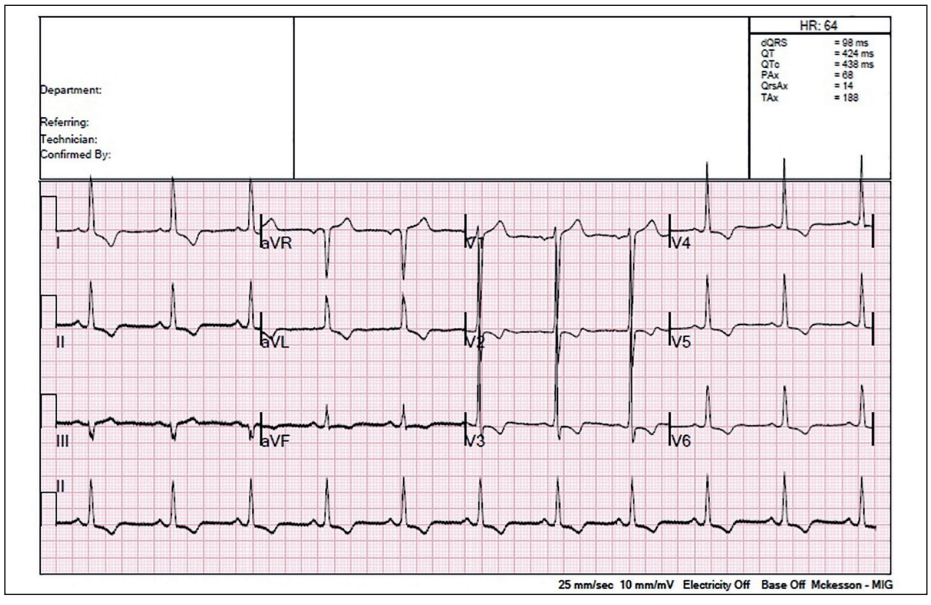
Figure 1. Electrocardiogram showing widespread repolarization abnormalities and evidence of left ventricular hypertrophy.



CASE REPORT

Mr B, a 38-year-old paramedic, presented with a 5-day history of intermittent central chest pain radiating to his neck with occasional palpitations and dizziness, on the background of exertional dyspnoea over the past year. He had suffered with a self-limiting viral upper respiratory tract infection 2 weeks previously. There was no previous history of childhood illnesses or past hospital visits.

He was an ex-smoker with a 15 pack-year history and otherwise had no further risk factors for ischaemic heart disease. He rarely drank alcohol. He played recreational aerobic sports on a regular basis and was able to keep up with the physical demands of his job. He had no family history of cardiac disease or unexplained sudden death.

On examination he was clinically euvolaemic with a blood pressure of 158/91 mmHg and heart rate of 85 beats per minute. The apex beat was undisplaced and there were no palpable heaves or thrills. His jugular venous pressure was measured at 3 cm above the sternal angle and heart sounds were normal with no added sounds. His chest was clinically clear on auscultation and his calves were soft and non-tender.

His electrocardiogram on admission (*Figure 1*) showed sinus rhythm with widespread non-dynamic anterolateral T wave inversion and evidence of left ventricular hypertrophy. There was an upright T wave in lead aVR and large positive QRS deflections in leads I and II. QRS duration and QTc were 102 ms and 454 ms respectively.

Chest X-ray revealed a globular central cardiac shadow at the upper limits of normal size and clear lung fields.

Initial transthoracic echocardiography performed locally was a technically difficult study with abnormal, non-standard windows. This demonstrated severe left ventricular impairment with severe mitral regurgitation and abnormal valve morphology. There was also a global pericardial effusion and an abnormal position of the aorta and aortic valve (*Figure 2*).

Subsequent cardiac magnetic resonance imaging allowed the conclusive diagnosis of congenitally corrected transposition of the great arteries (*Figure 3*).

Mr B was referred to the adult congenital heart disease service at a specialist tertiary centre for long-term follow up. He currently has minimal symptoms and practises a minimally strenuous lifestyle.

Figure 2. Transthoracic echocardiography showing atrioventricular discordance with apical displacement of the morphological tricuspid valve in the four-chamber view.

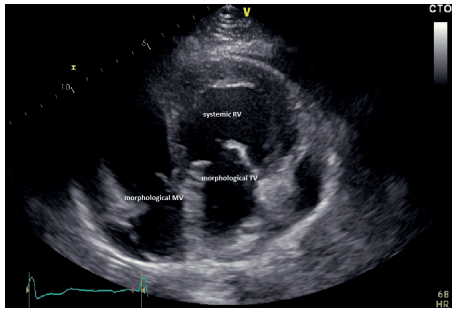
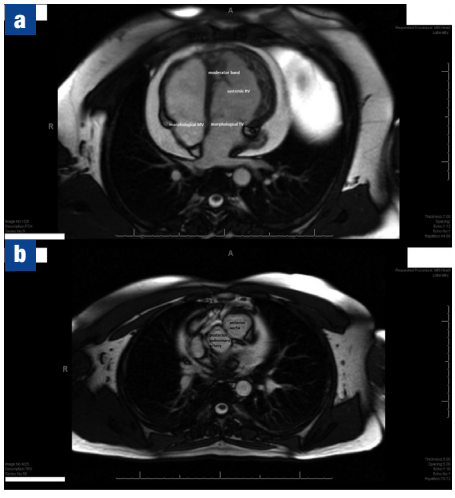


Figure 3. **a.** Morphology of the right ventricle is illustrated on cardiac magnetic resonance imaging in the four-chamber view demonstrating coarse trabeculations and a moderator band. **b.** The axial plane demonstrates an anteriorly positioned aorta, left of the pulmonary artery.



case, however, no other defects were found which may explain the clinical presentation later in life.

In a study of antenatal diagnoses, moderate or severe right ventricular dysfunction was displayed in 56% of those with associated defects compared to 32% without (Sharland et al, 2005). Patients with congenitally corrected transposition of the great arteries are more likely to develop congestive cardiac failure with advancing age and the majority will have developed this by their fifth decade (Graham et al, 2000).

Aerobic capacity is significantly diminished in these patients. A retrospective study of 41 patients with congenitally corrected transposition of the great arteries demonstrated reduced maximal oxygen uptake values on cardiopulmonary exercise testing in the group with congenitally corrected transposition of the great arteries, corresponding to 30–50% of the results achieved by healthy patients (Fredriksen et al, 2001).

Echocardiography remains the mainstay of diagnosis, although cardiac magnetic resonance imaging has taken on an increasing role, particularly in defining anatomy, and in functional and volumetric assessment of the systemic right ventricle (Schmidt et al, 2000; Teo and Hia, 2011). Gadolinium-enhanced magnetic resonance imaging in combination with cardiopulmonary exercise testing has previously been used by Giardini et al (2006) to demonstrate right ventricular myocardial fibrosis hypothesized to be responsible for right ventricular dysfunction.

Patients with congenitally corrected transposition of the great arteries have abnormal conducting system anatomy which makes them prone to developing arrhythmias. Holter monitoring, event recorder and electrophysiology testing are required for selected patients and many eventually require permanent pacing for complete heart block (Anderson et al, 1974; Baumgartner et al, 2010). The incidence of sudden cardiac death in congenital heart disease is approximately 1:1000 patients per year, although ventricular tachycardia has rarely been described in the natural history of congenitally corrected transposition of the great arteries (Harrison et al, 1996).

There is limited evidence for conventional medical management of ventricular dysfunction in the context of a systemic right ventricle. Corrective anatomical surgery is considered in childhood with the best outcomes in those under 10 years of age. All patients with congenitally corrected transposition of the great arteries should be followed up by a cardiologist with a special interest in congenital heart disease (Baumgartner et al, 2010; Hornung and Calder, 2010).

The differential diagnosis of cardiac symptoms coupled with an abnormal electrocardiogram in a young adult commonly includes premature coronary artery disease, hypertrophic cardiomyopathy, aortopathy, valvular heart disease and arrhythmia. This case illustrates the rarity of congenital heart disease presenting later in life and the importance of considering this disease group when faced with a common clinical scenario such as this. **BJHM**

LEARNING POINTS

- Congenitally corrected transposition of the great arteries can present in the fourth decade of life with congestive cardiac failure if not associated with another congenital defect.
- Patients with congenitally corrected transposition of the great arteries are prone to developing arrhythmias and many eventually require permanent pacing for complete heart block.
- There is limited evidence supporting the use of conventional medical therapy for ventricular dysfunction, in the context of the systemic right ventricle.
- Late presenting adult congenital heart disease is rare but must be considered in the differential diagnosis when a young patient presents with cardiac symptoms.

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