A 30 year-old man with history of heart murmur since childhood presented with progressive dyspnea for 2 years. He recalled having some problems participating in school days; nonetheless, it had been uneventful until recently when his exercise capacity began to decline. He could barely walk up one flight of stairs and reported intermittent feet swelling and symptoms of paroxysmal nocturnal dyspnea. His blood pressure was 115/75 mmHg, pulse 108/min and pulse oximetry of 88%. He appeared cyanotic and his fingers had mild clubbing appearance. He had parasternal, apical heaving without thrill and laterally-displaced apex. His S1 was normal with a loud P2. A faint systolic murmur was audible at the upper left parasternal area. His echocardiographic study revealed a large membranous ventricular septal defect (VSD)(Figure 1) with bi-directional shunt, right ventricular hypertrophy (Figure 2) and right ventricular (RV) systolic pressure of 120 mmHg. Cardiac magnetic resonance imaging (MRI) was performed to identify other congenital abnormalities and revealed a moderate-sized patent ductus arteriosus (PDA)(Figure 3), a markedly dilated pulmonary artery, dilated left ventricle (LV) with left ventricular ejection fraction (LVEF) of 41% and right ventricular ejection fraction (RVEF) of 45%.

Late gadolinium enhancement images revealed two triangular-shaped areas of hyperenhancement at both RV insertion points of the interventricular septum (Figure 4). Calculated shunt (Qp: Qs) with phase-contrast MR was 0.85 to 1, consistent with a balanced cardiac shunt. Cardiac catheterization confirmed the result of echocardiogram and MRI findings and the patient was recommended heart-lung transplantation. Meanwhile, he was started on Bosentan for palliation with substantial improvement of symptoms.
Discussion

This is an example of typical presentation of patient with Eisenmenger syndrome from congenital defects with left-to-right shunt such as atrial septal defect (ASD), VSD and PDA.

Standard investigations in these patients mainly include echocardiography and cardiac catheterization which often provide adequate information for diagnosis and management. Although most patients would likely be in the heart transplantation waiting list, only a small number of patients on the list would receive their new hearts and lungs. With limited number of heart transplantation, new therapies aiming to treat pulmonary hypertension and to improve morbidity and mortality are undergoing clinical studies.

With advance of cardiac imaging today, combination of echocardiographic study and cardiac MRI, instead of cardiac catheterization in many cases, can often give adequate anatomical and hemodynamic information of congenital cardiac anomalies in most complex congenital heart disease patients for complete diagnosis and planning therapy. The advantage of MRI, apart from being non-invasive and radiation-free method, LV/RV anatomy and function can be accurately and reliably evaluated and followed up after therapy by cardiac MRI which has become a gold standard for ventricular function assessment. MRI can also accurately assess flow rate and flow volume of any cardiac orifices and vessels, therefore cardiac output, regurgitant volume and cardiac shunt can be calculated. The disadvantages of MRI compared with invasive catheterization include direct pressure and oxygenation measurement which may preclude detection of small shunts.

Characteristic pattern of late enhancement MRI includes mid-myocardial hyperenhancement lesions along the RV insertion of interventricular septum, which are best visualized in short view of the ventricle, may appear at anterior or inferior insertion points or both. The late gadolinium enhancement finding in severe pulmonary hypertension, as in our patient, correlates well with pulmonary pressure and a potential tool for assessing prognosis and response to medical treatment for pulmonary hypertension and RV hypertrophy (1,2).

Figure 3. Axial black-blood T1-weighted image showing a patent ductus arteriosus (PDA) connected pulmonary artery and descending aorta

Figure 4. Short-axis delayed enhancement image demonstrates hyperenhanced lesions (arrows) at both RV insertion points, one of MRI features of severe pulmonary hypertension

References