INTRODUCTION

We present a case of a congenital absence of pericardium in a young adult presenting with chest pain. Discussions of the characteristic rest and stress echocardiography and electrocardiography (ECG) features are laid out. A brief description of typical ECG and chest radiograph findings is also provided.

CASE PRESENTATION

A 21-year-old Chinese man, physically active and with no known previous hospitalizations, was admitted to the Cardiology Department of Khoo Teck Puat Hospital for left-sided chest pain that was atypical in nature.

Physical examinations were unremarkable except for a displaced apical impulse. Blood tests were likewise unremarkable. Serial high sensitive troponin, creatinine kinase, and creatine kinase-MB were normal. ECG showed sinus bradycardia with a rate of 57 beats per minute, right axis deviation, right bundle branch block (RBBB) pattern, poor R wave progression, and early repolarization changes per minute, right axis deviation, right bundle branch block (RBBB) were normal. ECG showed sinus bradycardia with a rate of 57 beats per minute, right axis deviation, right bundle branch block (RBBB) pattern, poor R wave progression, and early repolarization changes.

We then sent the patient for an exercise stress echocardiography. Baseline echocardiography showed an unusual echocardiographic appearance: a right ventricle that seemed to be enlarged, a tear-drop-shaped heart (Figures 3–5 and Videos 1–3), a hypermobile heart, and a paradoxical septal motion of the interventricular septum on M mode (Figure 6).

Stress images showed a heart that seemed to be swinging in a pendulum manner (Videos 4 and 5). ECG at peak stress showed QRS complexes that changed continuously in axis or QRS alternans as shown by the arrows in Figure 7. We then diagnosed the patient as a case of congenital absence of pericardium. Further history taking revealed that the patient had a computed tomography (CT) scan of the thorax done a year prior for investigation of a possible lung emphysema in another hospital, which confirmed our diagnosis.

The CT thorax revealed a relative shift of the mediastinum to the left (Figure 8), an absent pericardium, and lung interposition between the aorta and pulmonary artery (Figure 9).

DISCUSSION

Congenital absence of pericardium was first clinically diagnosed in 1959 by Ellis et al.1 It is a rare condition that as of 2013 has been reported in only about 400 cases, and oftentimes the defect is discovered incidentally during operations or postmortem (1/10,000 autopsies).3

The diagnosis of congenital absence of pericardium requires a high degree of suspicion because the usual clinical presentations are nonspecific, and given the potential complications, diagnosis and detection are clinically relevant.4 Clinical presentations vary from chest pain to myocardial infarction, syncope, tricuspid regurgitation, and sudden death.5,6

Suspicion of this diagnosis initially arises on physical examination and on review of characteristic ECG and chest radiograph findings and is subsequently supported by echocardiography findings and confirmed by cardiac magnetic resonance imaging (CMR) or CT. Physical examination may reveal a laterally displaced apex beat, systolic murmurs, and clicks of undetermined origin.5

ECG findings are typical but not diagnostic. They may reveal bradycardia, right axis deviation, complete or incomplete RBBB, poor R wave progression throughout the precordial leads, and prominent P waves in the midprecordial leads. The poor R wave progression is usually secondary to lateral displacement of the precordial transition zone, and the prominent P waves may be due to right atrial overload.5,6

Classic features of the chest radiograph include leftward displacement of the heart, which is confirmed by the absence of the right heart border projecting on the right of the vertebral column; left cardiac border straightening and elongation (“Snoopy sign”); and a lucent area between the aorta and pulmonary artery caused by a deep or very well-defined aortopulmonary window, which is caused by the absence of pleura, allowing lung to invaginate into this space.5,6

Although CMR is the gold standard in the diagnosis of congenital absence of pericardium, echocardiography remains an important tool that can be utilized when this diagnosis is suspected as it can accurately identify any abnormalities in cardiac appearance, motion, and hemodynamics.5,6

The characteristic features seen in a resting echocardiogram are the pathognomonic teardrop-shaped heart with elongated atria and ventricles; unusual echocardiographic windows; cardiac hypermobility; cardiopatosis, which is the marked change in cardiac position with changing patient position on the examination table; abnormal swinging of the motion of the heart; paradoxical septal motion of interventricular septum on M mode, which can be explained by the apparent lack of restraint to the heart’s motion; a false-positive appearance of right cavity dilatation; and pulse Doppler pulmonary vein flow evaluation showing reduced systolic flow and systolic-to-diastolic flow ratio.3,6

Connolly et al. from 1982 to 1992 in the Mayo Clinic, studied 10 patients diagnosed with congenital absence of pericardium who
underwent echocardiography. Results showed that an unusual echocardiographic window was seen in all of the patients, cardiac hypermobility in nine patients, abnormal ventricular septal motion in eight patients, and abnormal swinging motion of the heart in seven patients.

During stress echocardiography, images may show an erratic pendulum-like movement of the heart, accompanied by continuous QRS axis change or QRS/electric alternans. This electric alternans is secondary to the exaggerated translational movement of the heart.

Topilsky et al. described this exaggerated pendulum movement of the heart during effort and the beat-to-beat morphological QRS changes during stress electrocardiography in 2010. These changes had previously been described only with large pericardial effusions or in those patients who had severe blunt trauma to the chest resulting in pericardial rupture.

Congenital absence of pericardium can be classified as partial left (70%), right (17%), or total bilateral (extremely rare) pericardial absence. It is three times more common in males than in females.

Congenital absence of pericardium, either complete or partial, usually has an excellent prognosis, although all types can occasionally lead to serious complications. Generally, complications are related to the type and size of the defect. Partial defects are likely susceptible to complications compared with complete bilateral absence.

The most commonly documented complications are herniation and incarceration of the myocardium, predominantly the left atrial appendage; however, herniation of the ventricles has also been reported. Torsion of the great vessels leading to aortic dissection can also happen, and it is secondary to increased cardiac mobility within the chest cavity. Coronary artery compression by the edge of the pericardial defect can lead to myocardial ischemia, and cardiac traction has been reported due to adherence of the epicardium to other chest structures in the absence of the pericardial and pleural membranes and due to valvular insufficiency.

Surgical treatment is recommended for patients who develop complications like imminent strangulation. Surgical options include left atrial appendectomy, division of adhesions, pericardiectomy, and pericardioplasty (Dacron, Gore-tex, or bovine pericardium). Pericardiectomy enlarges the defect to reduce the risk of incarceration. Pericardioplasty aims to restore the defect either by primary closure or complete reconstruction with synthetic materials. Postpericardiotomy syndrome is a common complication following these surgeries.

In those patients who present with no complications, various strategies are proposed. One proposed approach is to estimate the risk of life-threatening complications based on the size of the pericardial defect. Total left defects usually warrant no surgical treatment because they usually carry a very low risk of herniation.

The management approach for left partial pericardial defects, on the other hand, is more complex and controversial. Some practitioners argue that all partial pericardial defects are high risk, irrespective of defect size, for developing complications, while others divide the risk.

Figure 1 ECG showing bradycardia, RBBB pattern, and poor R wave progression.
according to the size of the defect. With respect to defect size, due to their low risk of complications compared with total defects, asymptomatic, large partial pericardial defects are not surgically treated.2,5 On the other hand, diagnosis of a small to moderate-sized pericardial defect in a symptomatic or asymptomatic patient is considered by some practitioners as a cause for prophylactic operation to reduce the risk of death from cardiac structure herniation and incarceration, while others suggest surgery only for symptomatic patients.5 Overall, in asymptomatic patients, due to the good to excellent prognosis, follow-up and watchful waiting will suffice.5

For the present case, as our patient remained asymptomatic during his stay in the hospital and no complications were seen in connection with his absent pericardium, he was discharged with regular follow-up for observation.
CONCLUSIONS

Congenital absence of pericardium is a rare defect of which recognition is critical because it can be associated with catastrophic outcomes. Symptomatology is nonspecific and requires a high degree of suspicion. Diagnosis should be considered when characteristic ECG, chest radiograph, and echocardiography findings are present. Although CMR is the gold standard in the diagnosis, echocardiography remains an important tool in assessment when suspicion of absent pericardium is present. Stress echocardiogram with findings of any abnormal echocardiographic and electrocardiographic responses is a useful tool and can aid in the diagnosis. The diagnosis entails a good to excellent prognosis for asymptomatic patients and may only require watchful waiting. Surgery, on the other hand, is recommended for symptomatic patients with complications.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at http://dx.doi.org/10.1016/j.case.2017.01.004.

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Figure 9  CT scan showing lung interposition between the aorta and pulmonary artery (arrows) and relative shift of mediastinum to the left and an absent pericardium.