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CASE REPORT
Congenital heart disease

Diagnosis and management of congenital absence of pericardium: a case report

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Background Absence of the pericardium is a rare congenital defect with an approximate incidence of <1/10 000. We review a case of complete pericardial agenesis in a symptomatic patient with gross cardiac mobility, for which pericardial reconstruction was undertaken successfully.

Case summary A 24-year-old otherwise fit and well patient, with debilitating exertional chest pain was found to have complete pericardial agenesis on the left side and on the diaphragmatic surface. There was gross cardiac mobility demonstrated on cardiac magnetic resonance imaging. His pericardium was reconstructed surgically using Gore-tex® patches. There were no complications, and the patient was discharged 8 days later. Three months later at follow-up, the patient required no analgesia and has had complete resolution of his chest pains.

Discussion Congenital hemi-pericardial agenesis is a very rare condition which often remains undetected due to its asymptomatic nature. It is important to consider this as a differential diagnosis of exertional chest pains. Cardiac magnetic resonance imaging remains the investigation of gold standard. There is no consensus on whether surgical intervention in symptomatic or asymptomatic patients has any prognostic value. However, we have demonstrated that by reconstructing the pericardium in a highly symptomatic patient, there has been a resolution in size of a previously dilated right ventricle and most importantly an improvement in quality of life.

Keywords Pericardium • Agenesis • Absent • Reconstruction • Congenital • Case report

Learning points

- Congenital absence of pericardium is an exceedingly rare condition which may remain undetected due to lack of symptoms.
- Consider it as a differential diagnosis of atypical, angina-like pain with unobstructed coronary arteries and evidence of cardiac mobility.
- Clinical examination and Chest X-ray (CXR) often show leftward displacement of the heart. Cardiac magnetic resonance imaging is the gold standard investigation.
- There is no consensus on management, albeit severely symptomatic patients should be considered for surgical reconstruction aiming for symptomatic relief.

Introduction

Absence of pericardium is a rare congenital anomaly, with an approximate incidence of <1/10 000.[1,2] This may be an underestimate, due to either a lack of symptoms or their non-specific nature.

The role of the pericardial sac is to anchor and stabilize the heart within the thoracic cavity, and the serous fluid reduces friction.[3] The pericardium also acts as a physical barrier to infection and to trauma.[4]

Complete left-sided pericardial agenesis occurs in 70% of cases, right-sided absent pericardium in 17% followed by complete bilateral in 9% of cases.[5]

Pericardial agenesis may be asymptomatic or associated with atypical chest pains thought to be due to cardiac mobility or torsion.[6]

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Some also report breathlessness on lying to one side—a phenomenon called trepopnoea which is uniquely linked to partial defects.[7]

Up to 50% of patients with pericardial agenesis have an associated congenital heart defect. These include atrial septal defects, sinus venosus defects with partial anomalous pulmonary venous drainage, tetralogy of Fallot, patent ductus arteriosus and mitral valve disease.[8]

In complete left-sided pericardial agenesis, the apex beat is displaced to the left and there may be a systolic murmur at the left sternal which is thought to be due to turbulent flow caused by a hypermobile heart.[5] Poor R wave progression on electrocardiogram and change in axis could reflect the left-sided displacement and torsion of the heart.[3]

Chest X-rays tend to show a leftward and posterior shift of the cardiac outline, with a straight left heart border and effacement of the right.[9] Echocardiography may show increased cardiac motion, and a dilated right ventricle (RV). This is due to cardiac chambers being shifted to the left.[10] On cardiac computed tomography (CT), lung tissue may be seen in between the aorta and the pulmonary artery and in between the base of the heart and the diaphragm.[6]

We review the case of an absent pericardium which was successfully surgically reconstructed. The focus of this report is, however, on how to evaluate and manage symptomatic patients.

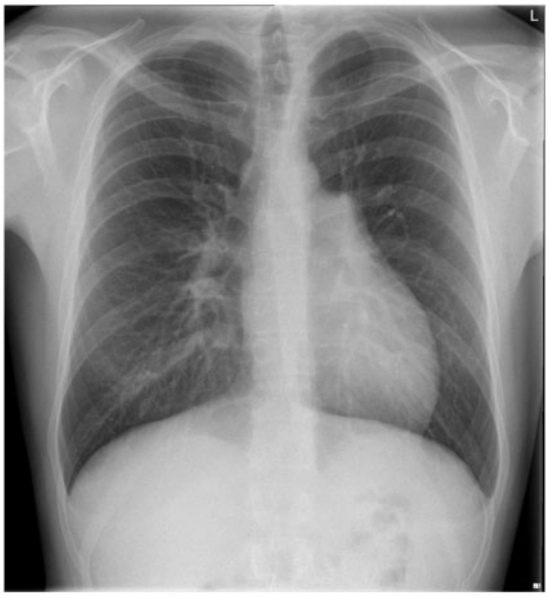


Figure 1 Chest X-ray.

Timeline

June 2016	Patient complains of atypical left-sided chest pains and is referred to cardiology
May 2018	Imaging confirms complete absence of left pericardium on cardiac magnetic resonance imaging
June 2018	Discussed at Adult Congenital Heart Disease Multidisciplinary Team (MDT) at The Royal Brompton Hospital
August 2018	Admission for surgical reconstruction of pericardium
	Discharged Day 9 with improving symptoms
November 2018	Follow-up clinic: patient symptom-free

Case presentation

A 24-year-old gentleman with no past medical history had been experiencing debilitating chest pains for 2 years. He described these as varying in intensity, worsened by exercise, and relieved partially by analgesia. They were not associated with palpitations or syncope.

Unfortunately, due to his symptoms he had to give up his studies and minimize his physical activity. He had trialled multiple analgesics (daily paracetamol and tramadol) with partial relief of symptoms.

He underwent serial investigations at his local cardiology unit, including a cardiac CT which identified an absence of pericardium on the left side of the heart. He was referred to the Adult Congenital Heart Disease (ACHD) team at The Royal Brompton Hospital.

When first reviewed in the ACHD clinic he was not cyanosed, in sinus rhythm with no evidence of cardiovascular decompensation. His apex beat however was grossly displaced to the left and more so on lying supine. His first heart sound was normal, second split, there was also a soft one-sixth systolic heart murmur. The rest of his examination was unremarkable.

His electrocardiogram showed him to be in sinus rhythm at 78 b.p.m., with normal conduction times but a poor R wave progression and an intermittently changing axis.

A chest radiography (*Figure 1*) which showed situs solitus, levocardia, with a left aortic arch, no displacement of the heart to the left, and a protrusion at the level of the left atrial appendage at the left upper quadrant of the heart.

His echocardiogram showed a structurally normal heart with maintained ventricular function. Stress echocardiography showed mild right ventricular dilatation at rest with increase in right ventricular volume directly after exercise. A cardiac CT (*Figure 2*) revealed a partial absence of pericardium on the left side with a small layer of pericardium on the upper right heart border. The heart was grossly shifted leftwards and posteriorly, highly suggestive of enhanced heart mobility. Similarly, no pericardium could be seen on cardiac magnetic resonance imaging (CMR). There was lung tissue in between the base of the heart and diaphragm (*Figure 3A, B, E*), the heart was displaced to the left of the chest and the RV appeared dilated (*Figure 3C, D*).

The patient's symptoms were thought to be due to his absent pericardium and related to cardiac mobility. We were struck by the discrepancy of the erect Chest X-ray (CXR) and supine CMR with regards to cardiac position, mobility, and the severity of disabling symptoms. We thus discussed the patient at our ACHD Multidisciplinary Team meeting and offered him surgery, in an attempt to immobilize his heart, hoping to improve his symptoms and quality of life.

Reconstruction of the pericardium

The heart was exposed via a left postero-lateral thoracotomy. There was complete pericardial agenesis on the left side and on the diaphragmatic surface. Several 0.4 mm thickness Gore-tex® patches were sutured together to create a large sheet and this was sutured down with widely spaced interrupted 3-0 Prolene® sutures to the posterior mediastinum, starting at the level of the left pulmonary artery, posterior to the left atrial appendage and along the left hilum and inferior pulmonary ligament, to the diaphragm and down. This was then brought forward to the anterior chest wall and then fixed anteriorly along the anterior chest wall with the superior end free (see *Figure 4*). A left pleural drain was inserted and a paravertebral catheter for analgesia, followed by routine multilayered chest closure.

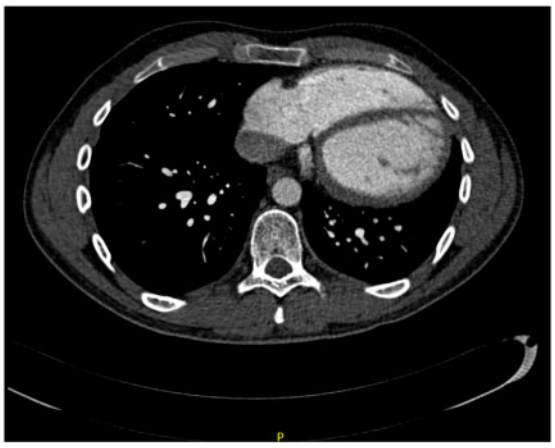


Figure 2 Computed tomography scan pre-operative.

The post-operative course was uneventful apart from a small left apical pneumothorax with a small pleural effusion. Both resolved spontaneously within days.

Echocardiography on Day 5 showed that the RV was of normal size with no evidence of pericardial effusion nor compression of cardiac chambers. The patient was discharged 8 days after his procedure on simple analgesia, mobilizing well with improved symptoms.

Three months from surgery the patient had come off analgesia completely, with resolution of his debilitating chest pains. He was resuming his University studies and restoring normality in his life.

Discussion

There seems to be no real consensus on how to manage patients with pericardial agenesis. A subgroup of partial defects is associated with cardiac strangulation and coronary artery compression which may lead to sudden death.[11,12] These patients, if symptomatic, may

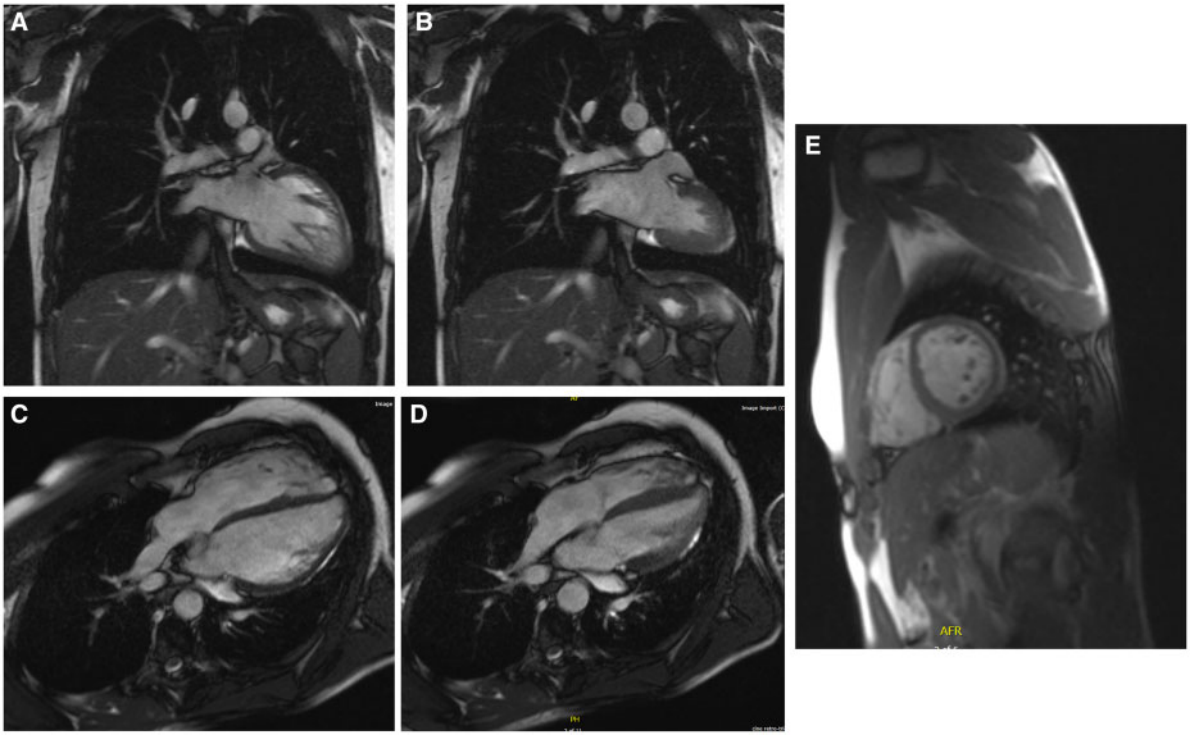


Figure 3. (*A*) Pre-operative cardiac magnetic resonance imaging coronal cine diastole. (*B*) Pre-operative cardiac magnetic resonance imaging coronal cine systole. (*C*) Pre-operative cardiac magnetic resonance imaging cine four chamber view diastole. (*D*) Pre-operative cardiac magnetic resonance imaging cine four chamber view systole. (*E*) Pre-operative cardiac magnetic resonance imaging sagittal trufi short-axis view.

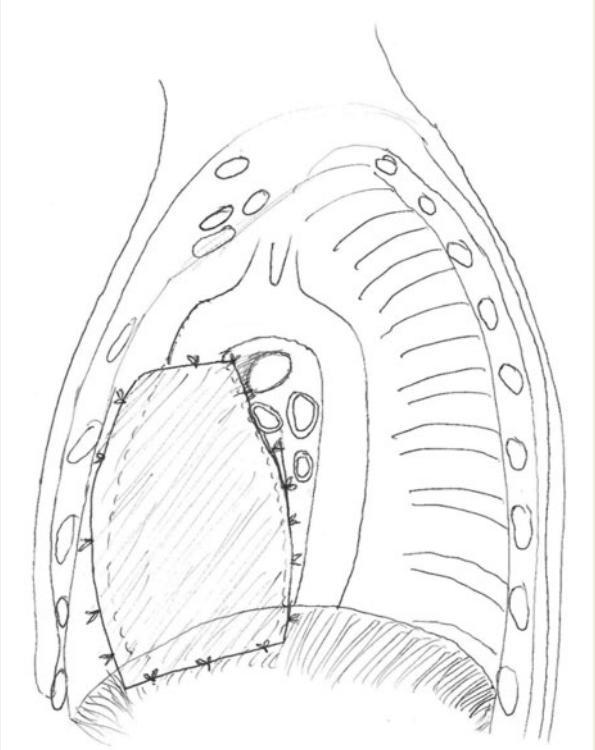


Figure 4 Schematic of the position of the ePTPF patch sutured into position through a left thoracotomy to resuspend the heart in a more natural position and prevent further cardiac mobility. Hoschtitzky A. Reconstruction of the pericardium. 2019.

be offered surgery.[5,6] They may initially present with chest pain and ST elevation. Coronary angiogram in such cases will tend to reveal normal coronaries with an abrupt kink due to compression by the pericardial rim.[12] The left atrium, left atrial appendage, and regions of the left ventricle can herniate through partial defects. These can be identified on echocardiogram and CMR.[13] If those defects are found incidentally in asymptomatic patients the management causes debate. Some would offer surgery to avoid complications from herniation or strangulation.[14] Others would suggest observing and monitoring asymptomatic patients.[15]

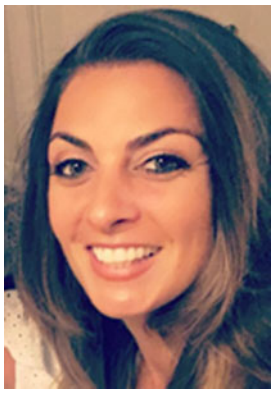
In a review[6] looking at four patients with total absence of the pericardium, who were symptomatic and underwent surgery, it was found that all reported significant improvement in severity and frequency of symptoms post-operatively, and two of them became symptom-free. Such study would thus imply that pericardioplasty in symptomatic patients with complete agenesis of the pericardium, with low morbidity, would benefit from surgical intervention. This mirrors the experience that we have had with our patient.

Conclusion

Congenital absence of pericardium is an exceedingly rare condition which may remain undetected due to lack of symptoms. However, it is important to consider it as a differential diagnosis of atypical, angina-like pain with unobstructed coronary arteries, and evidence of cardiac mobility. Clinical examination and CXR often show leftward displacement of the heart, whereas CT and CMR confirm the diagnosis (the latter being the gold standard investigation). There is no consensus on management, albeit severely symptomatic patients should be considered for surgical reconstruction aiming for symptomatic relief.

We have demonstrated that by using a technique to reconstruct the pericardium in a highly symptomatic patient, there has been a resolution in size of a previously dilated RV and most importantly an improvement in quality of life.

Lead author biography



Dr Melissa Bouchard trained in Manchester Medical School and completed her general cardiology training in the North West Deanery. She is currently subspecializing in adult congenital heart disease at The Royal Brompton Hospital in London.

Supplementary material

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

Slide sets: A fully edited slide set detailing this case and suitable for local presentation is available online as Supplementary data.

Consent: The author/s confirm that written consent for submission and publication of this case report including image(s) and associated text has been obtained from the patient in line with COPE guidance.

Conflict of interest: none declared.

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