Cardiac Eclipse: Congenital Absence of the Pericardium Manifesting as Atypical Chest Pain

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INTRODUCTION

Congenital absence of the pericardium (CAP) is a rare abnormality that results from a failure of the pleuropericardial membranes to fuse completely on one or both sides. Because it is rare, clinicians struggle both with recognition and with accurate diagnosis. We present the case of a 29-year-old man with chest pain who was incidentally discovered to have CAP.

CASE PRESENTATION

A 29-year-old man with a medical history of hyperlipidemia presented as a new patient visit to an outpatient cardiology office. He had recently visited an emergency department twice on consecutive days for evaluation of atypical chest pain. This was described as left-sided, radiating up his left jaw, six out of 10 in intensity, not associated with exertion, lasting 15 min, occurring several times per month and abating on its own without the use of any pain medications. On both visits, his workup was reported as unremarkable, including chest radiography (CXR) described as normal; a basic metabolic panel with normal electrolytes and kidney function; a complete blood count without leukocytosis, anemia, or thrombocytopenia; an undetectable troponin I level; and negative urine drug screen. Electrocardiography showed sinus bradycardia, and he was told to establish care with cardiology.

In the office, the patient’s blood pressure was 116/80 mm Hg, and his heart rate was 47 beats/min. On physical examination, he was noted to have an irregular rhythm but no murmurs, rubs, or gallops. His point of maximal intensity was not displaced. His electrocardiography (CXR) described as normal; a basic metabolic panel with normal electrolytes and kidney function; a complete blood count without leukocytosis, anemia, or thrombocytopenia; an undetectable troponin I level; and negative urine drug screen. Electrocardiography showed sinus bradycardia, and he was told to establish care with cardiology.

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Cardiac computed tomographic (CT) imaging confirmed the diagnosis of congenital absence of the left pericardium with minimal residual right-sided pericardium (Figures 3 and 4). CT imaging also demonstrated interposition of lung tissue between the heart and diaphragm and between the aorta and the main pulmonary artery (PA). On reconstructed helical CT images, a portion of the acute marginal artery coming off the right coronary artery appeared to be impinged by the residual right-sided pericardium, which would be considered a high-risk feature (Figure 5). No additional structural abnormalities were noted on the CT scan. Given the possibility of coronary impingement noted on CT imaging, a myocardial perfusion study with nuclear imaging was performed. The gated images demonstrated normal contraction of all myocardial segments, with no evidence of ischemia. When outside CXR done at the initial emergency department visit became available for review, findings typical of CAP were in fact noted that had been missed, including Snoopy’s sign and levorotation of the heart, as well as radiolucency between the aortic knob and the main PA, with loss of the right heart border (Figure 6).

The patient was seen in follow-up several months later and reported complete resolution of his atypical chest pain. Although there was initial concern for a high-risk feature with possible coronary artery impingement, his stress test did not demonstrate any ischemic changes; he was asymptomatic at peak exercise, and nuclear imaging did not demonstrate any regional ischemia. Surgical intervention was not indicated, and although he was offered the opportunity for referral to a center of excellence in pericardial diseases, he chose to follow up with his local cardiologist. Repeat imaging will be considered only for new and concerning electrocardiographic changes associated with clinical symptoms.

DISCUSSION

The spectrum of defects of CAP includes absence of the entire pericardium, absence of the left or right pericardium, a foramen-type defect of the left or right pericardium, or absence of the diaphragmatic pericardium. The distinction of these malformations is especially important because patients with partial defects are more likely to be asymptomatic, to have high-risk features, and to present with cardiac complications. Left-sided defects are most common (70%), followed by diaphragmatic defects (17%), bilateral or complete defects (9%), and right-sided defects (4%).

Although CAP usually exists as an isolated anomaly, up to 50% of patients may have additional cardiac and noncardiac defects. It has been linked to sinus venosus–type atrial septal defect with partial anomalous pulmonary venous drainage, tetralogy of Fallot, simple secundum atrial septal defects, and patent ductus arteriosus. On the basis of several recent case series and autopsy reports, the incidence of CAP ranges from 0.007% to 0.044%, with a male/female ratio of 3:1.
Because of both its rarity and usually silent presentation, CAP is typically diagnosed incidentally on imaging, during surgery, or postmortem. Symptoms include atypical chest pain, which may be positional in nature, occurring more often in the left lateral decubitus position, shortness of breath, palpitations, dizziness, arrhythmias, and syncope; rarely there is sudden cardiac death. Chest pain may occur because of a lack of pericardial cushioning, torsion of the great vessels, compression of coronary arteries, or tension from pleuropericardial adhesions. Positional chest pain is thought to arise from a lack of pericardial tissue, which allows more cardiac mobility.

Dyspnea and trespnea, or dyspnea that occurs while in the lateral decubitus position on one side but not the other, may occur as a result of lower left pulmonary vein compression between the left atrium and descending aorta. In patients with partial defects, it is possible for the heart to herniate through the remaining pericardial tissue and cause ischemia. Although herniation of the entire left atrium and both ventricles has been reported, the most common site of herniation is the left atrial appendage. Sudden cardiac death is the most feared complication and occurs because of cardiac tissue strangulation and compression of coronary vessels through partial foramen–type defects.

Diagnosis of CAP is made on the basis of abnormalities on physical examination, electrocardiography, CXR, and TTE imaging and confirmed by cardiac CT imaging or cardiac magnetic resonance imaging (CMR). Physical examination findings can include bradycardia, irregular heart rhythm, apical displacement of the point of maximal intensity (although our patient had a normal point of maximal intensity), and a systolic ejection murmur at the left sternal border, thought to be due to turbulence caused by a highly mobile heart. Changes commonly seen on electrocardiography include right-axis deviation, incomplete or complete right bundle branch block, and bradycardia from vagal nerve stimulation. Poor R-wave progression may also be seen because of leftward displacement of the transitional zone of the precordium. Coronary artery compression through a herniated segment of myocardium in partial-type defects can present with ST-segment elevation. Typical findings on CXR include levorotation of the heart, elongation of the left ventricular contour (Snoopy’s sign), radiolucency between the aortic knob and main PA, loss of the right heart border, and herniation of the left atrial appendage represented as a radiopaque bulge of the left upper heart border.

Perhaps the most appreciated echocardiographic finding of CAP is the unusual imaging planes necessary to obtain images. TTE features of CAP include posterior orientation of the apex, cardiopathy (a term first used in 1918 by Gordon in the British Medical Journal to describe “undue laxity of the cardiac supports,” currently used when there is evident downward displacement of the heart) in the parasternal long-axis view, with accentuated movement of the posterior left ventricular wall, and abnormal interventricular septal motion on M-mode imaging (“unrestrained heart”).
Excessive motion of the heart may be noted in all views. Dilatation of the right ventricle observed with far lateral positioning of the TTE probe, elongated atria with widened ventricles (teardrop appearance) resulting in an abnormal atrial-ventricular angle, and tricuspid regurgitation due to annular dilatation or chordal rupture may be operant.1–6

Once there are echocardiographic findings suggesting CAP, CT imaging or CMR should be performed to further characterize the pericardial defect and to identify any other associated lesions. CT imaging and CMR are comparable in diagnostic accuracy but offer distinct advantages, and selection depends on the clinical scenario, operator and interpreter expertise, and institutional availability. The advantage of CMR is that it provides a functional cardiac assessment, while cardiac CT can provide an assessment of coronary anatomy, which becomes crucial when dealing with partial-type defects that might result in coronary artery impingement and ischemia. Irrespective of modality, it is possible to visualize levorotation of the heart and interposition of lung tissue in areas of absent pericardium between the aorta and PA and between the diaphragm and the base of the heart. These findings are relatively specific for CAP.7
Direct confirmation of absence of the pericardium, presence of any residual pericardium and presence of coronary artery impingement is also possible. Importantly, any additional congenital cardiac malformations can be detected.

The rarity of CAP has precluded the establishment of management guidelines, and as a result, treatment recommendations are based largely on observational studies and case reports. Because the majority of cases are clinically silent, most patients are managed conservatively with reassurance, while surgery is reserved for those with high-risk features or unrelenting symptoms. Although most cases are found incidentally, it is important for clinicians to recognize the constellation of abnormal clinical and imaging findings for accurate anatomic diagnosis and risk stratification.

CONCLUSION

CAP is a rare congenital abnormality that can present with devastating cardiac consequences. The majority of cases are clinically silent and managed conservatively with reassurance, while surgery is reserved for those with high-risk features or unrelenting symptoms. Although most cases are found incidentally, it is important for clinicians to recognize the constellation of abnormal clinical and imaging findings for accurate anatomic diagnosis and risk stratification.

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SUPPLEMENTARY DATA

Supplementary data related to this article can be found at https://doi.org/10.1016/j.case.2019.07.005.

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