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*Case Report*

# Congenital Pericardial Agenesis: Innocent Finding or Clinically Significant Condition? A Case Series and Literature Review

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**Abstract**

Congenital pericardial agenesis (CPA) is a rare anomaly that is often considered a benign incidental finding but may present with nonspecific symptoms and mimic structural heart disease. Its clinical relevance remains incompletely defined, particularly regarding the distinction between harmless anatomical variant and clinically significant condition. We present a retrospective two-center case series of four patients with imaging-confirmed CPA, combined with a narrative review of the literature aiming to evaluate the clinical spectrum, diagnostic challenges, and management implications of CPA. The clinical presentation of our patients was heterogeneous, ranging from incidental findings to chest discomfort and dyspnea. In all cases, initial echocardiography suggested alternative diagnoses, including right ventricular cardiomyopathy, atrial septal defect, or pericardial disease, leading to diagnostic uncertainty. Definitive diagnosis was established using multimodality imaging, particularly cardiac magnetic resonance and computed tomography, which demonstrated characteristic features such as cardiac levoposition and interposition of lung parenchyma. Three patients had complete left pericardial agenesis and one had a partial defect. All patients were managed conservatively, with no complications observed during follow-up.

**Keywords:** congenital pericardial agenesisis; clinical relevance; treatment; follow-up

## 1. Introduction

Congenital pericardial agenesis (CPA) is a rare developmental anomaly characterized by complete or partial absence of the pericardial sac, with an estimated prevalence of 0.002–0.004% in the general population [1–3]. It results from defective embryologic development of the pleuropericardial membranes, most commonly due to premature atrophy of the left common cardinal vein (duct of Cuvier), leading to incomplete fusion during early gestation [2,4]. As a consequence, the heart lacks its normal fibrous restraint and may shift freely within the thoracic cavity, most often toward the left hemithorax [2,5,6].

CPA encompasses a spectrum ranging from complete absence of the pericardium—most frequently left-sided—to partial defects, which are considerably less common but clinically more significant. In the majority of cases, particularly in complete agenesis, the condition remains asymptomatic and is detected incidentally during imaging or surgical procedures performed for unrelated indications [5,7,8]. However, the absence of pericardial support may lead to characteristic anatomical and functional alterations, including cardiac levoposition, abnormal cardiac rotation, and interposition of lung tissue between mediastinal structures [5,9,10].

Despite its often benign course, CPA may present with nonspecific clinical manifestations such as chest pain, dyspnea, palpitations, or reduced exercise tolerance, frequently leading to misdiagnosis as structural heart disease or pericardial pathology [5,11,12]. Importantly, partial pericardial defects

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carry a risk of potentially life-threatening complications, including cardiac herniation, strangulation of cardiac chambers or appendages, and compression of coronary arteries, which may result in myocardial ischemia or sudden cardiac death [13–15]. These risks highlight the clinical relevance of distinguishing between complete and partial forms of the disease [8,16,17].

The diagnosis of CPA remains challenging due to the nonspecific nature of routine investigations such as electrocardiography, chest radiography, and transthoracic echocardiography [8,14,16]. Multimodality imaging plays a pivotal role, with cardiac computed tomography (CT) and particularly cardiac magnetic resonance (CMR) considered the gold standard for definitive diagnosis, allowing precise anatomical characterization and exclusion of differential diagnoses [5,8,14].

Given the rarity of the condition and the predominance of isolated case reports in the literature, the clinical significance of CPA remains incompletely defined, particularly regarding its distinction as a benign incidental finding versus a condition with potential clinical consequences. In this context, we present a series of four patients with congenital pericardial agenesis alongside a narrative review of the literature, aiming to clarify whether this rare anomaly represents an innocent incidental finding or a clinically relevant condition with potential implications for patient management.

## 2. Materials and Methods

### *2.1. Study Design and Case Selection*

This study represents a two-center case series combined with a narrative literature review. Four patients diagnosed with CPA were identified during routine clinical practice and imaging evaluation at tertiary cardiology and radiology centers in Sofia, Bulgaria. The cases were collected retrospectively based on the availability of complete clinical and imaging data.

All patients included in this series had a confirmed diagnosis of CPA established by advanced cardiac imaging modalities. Clinical data were obtained from medical records, including patient history, physical examination findings, instrumental and laboratory data, and follow-up information

### *2.2. Diagnostic Evaluation*

All patients underwent a stepwise diagnostic evaluation. Initial assessment included clinical examination, electrocardiography (ECG), routine laboratory investigations and transthoracic echocardiography (TTE), which in several cases raised suspicion for structural cardiac abnormalities but did not allow definitive diagnosis.

Definitive diagnosis was established using multimodality imaging, including contrast-enhanced cardiac computed tomography (CT) with ECG synchronization and cardiac magnetic resonance imaging (CMR). Characteristic imaging findings used to confirm CPA included:

- absence of the pericardium, most commonly along the left cardiac border;
- marked leftward displacement (levoposition) and rotation of the heart;
- interposition of lung parenchyma between the ascending aorta and pulmonary artery;
- separation of the heart from the diaphragm;
- absence of pericardial effusion despite apparent “free” cardiac contour.

These features were consistently demonstrated across cases. For example, CT imaging revealed absence of the left pericardium with lung interposition between the great vessels and adjacent to the left ventricular wall, as well as cardiac rotation with the apex directed upward. CMR further confirmed the absence of the pericardial layer and allowed detailed assessment of cardiac morphology and function, which remained preserved in all patients

### *2.3. Data Collection and Analysis*

Clinical and imaging data were systematically reviewed and analyzed. The following variables were collected for each patient:

- demographic characteristics (age, sex),

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- clinical presentation and symptoms,
- ECG findings,
- TTE findings,
- imaging characteristics on CT and/or CMR,
- type of pericardial defect (complete or partial),
- management strategy and clinical outcome.

Given the descriptive nature of this study and the small number of cases, no statistical analysis was performed. The cases were analyzed qualitatively to identify common clinical and imaging patterns.

### *2.4. Literature Review*

A narrative literature review was conducted to contextualize the findings of the presented cases. A structured search of the PubMed and Scopus databases was performed using combinations of the following keywords: "congenital pericardial agenesis", "pericardial absence", "partial pericardial defect", "congenital pericardial anomaly", "congenital pericardial malformation" and "cardiac herniation".

Articles published in English and reporting adult patients with imaging-confirmed CPA were considered eligible. Priority was given to case reports, case series, and review articles describing clinical presentation, imaging findings, complications, and management strategies. Relevant references from selected articles were also screened to ensure comprehensive coverage of the topic.

### *2.5. Ethical Considerations*

Our research was conducted in accordance with the principles of the Declaration of Helsinki. Patient consent was waived due to the retrospective and non-interventional nature of this case series and the use of fully anonymized clinical data, in accordance with applicable national regulations and institutional policies.

No artificial intelligence has been used for preparation of this paper.

## 3. Results

### *3.1. Case Series*

A total of four patients diagnosed with CPA were included in this case series. The clinical presentations ranged from incidental findings during imaging to evaluation for suspected structural heart disease. In all cases, the diagnosis was confirmed using advanced imaging modalities, primarily CT and/or CMR.

*Clinical case 1*

A 54-year-old male presented with progressive exertional dyspnea and fatigue, without associated chest pain, syncope, or palpitations. In February 2025, he experienced an episode of acute respiratory distress requiring short-term hospitalization, although no definitive cardiopulmonary cause was identified at that time. His medical history was notable for hypothyroidism, adequately controlled with levothyroxine, and dyslipidemia treated with low-dose atorvastatin.

On physical examination, the patient was in good general condition. Vital signs were within normal limits (heart rate 78 bpm, blood pressure 120/70 mmHg, respiratory rate 16/min, $SpO_2$ 98% on room air). Cardiovascular examination revealed regular heart rhythm and a soft holosystolic murmur at the cardiac apex. No peripheral edema or signs of heart failure were present. Laboratory investigations, including NT-proBNP (36.1 pg/mL), were within normal limits, effectively excluding decompensated heart failure.

Electrocardiography demonstrated sinus rhythm with left axis deviation and incomplete right bundle branch block, without ischemic changes (Figure 1).

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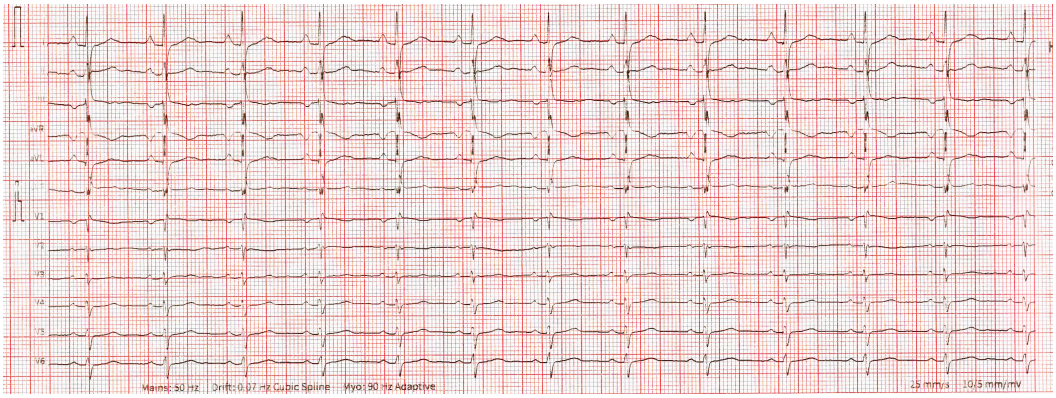

**Figure 1.** ECG of Clinical Case 1: Sinus rhythm (heart rate 75 bpm) with right axis deviation (+120°). Low QRS voltage in the precordial leads with leftward displacement of the R/S transition zone. Incomplete right bundle branch block pattern. Right ventricular hypertrophy cannot be excluded.

TTE was technically challenging due to the marked leftward displacement of the heart, resulting in poor acoustic windows and limited visualization. Standard parasternal and apical views could not be reliably obtained, and image interpretation was significantly restricted. Only subcostal views provided satisfactory image quality. From these, ventricular wall thickness was assessed as normal for both the left and right ventricles, with preserved left ventricular dimensions and systolic function, including normal ejection fraction estimated by the Teichholz method (Figures 2a, 2b, 2c)

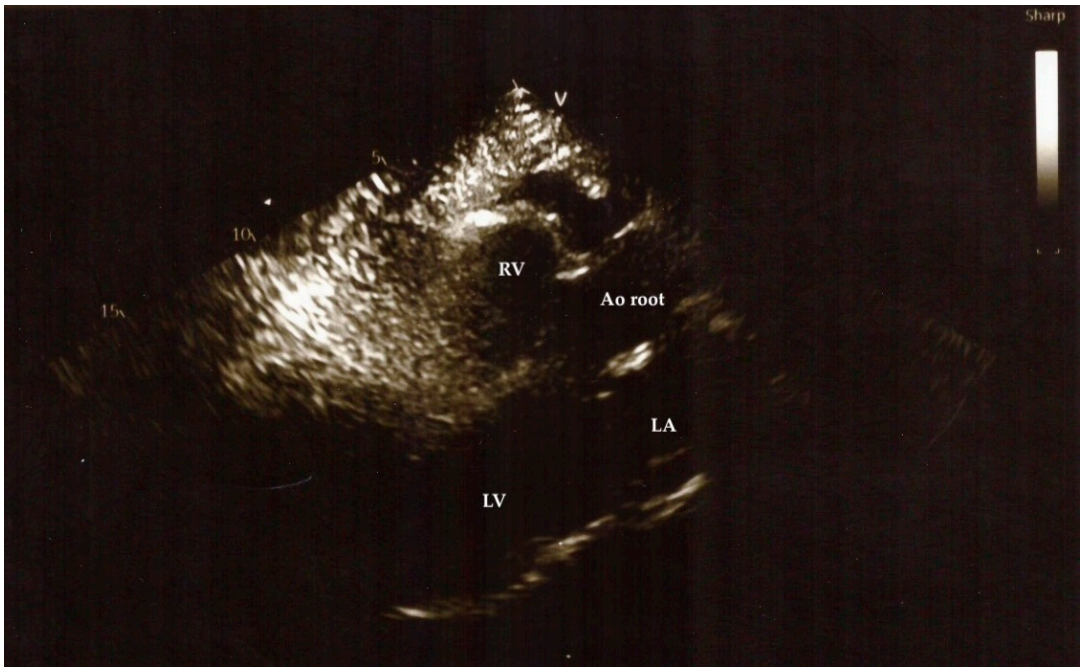


**Figure 2a.** TTE of Clinical Case 1: The parasternal long-axis view is suboptimal due to marked cardiac levoposition with interposition of aerated lung tissue, precluding adequate visualization and reliable assessment of cardiac structures and function from this window; Ao – aortic; LA – left atrium; LV – left ventricle; RV – right ventricle.

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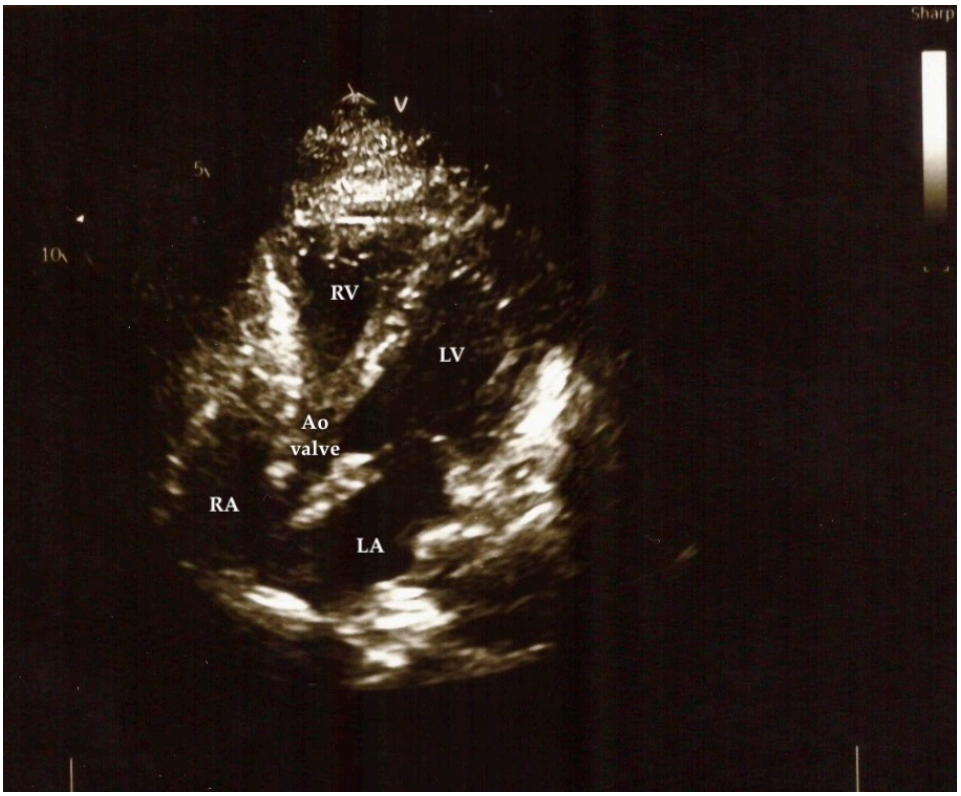


**Figure 2b.** TTE of Clinical Case 1: An apical five-chamber view was the only view obtainable from the apical window and was of suboptimal quality despite maximal left lateral positioning of the transducer. Standard apical four-, two-, and three-chamber views could not be acquired. Ao – aortic; LA – left atrium; LV – left ventricle; RA – right atrium; RV – right ventricle.

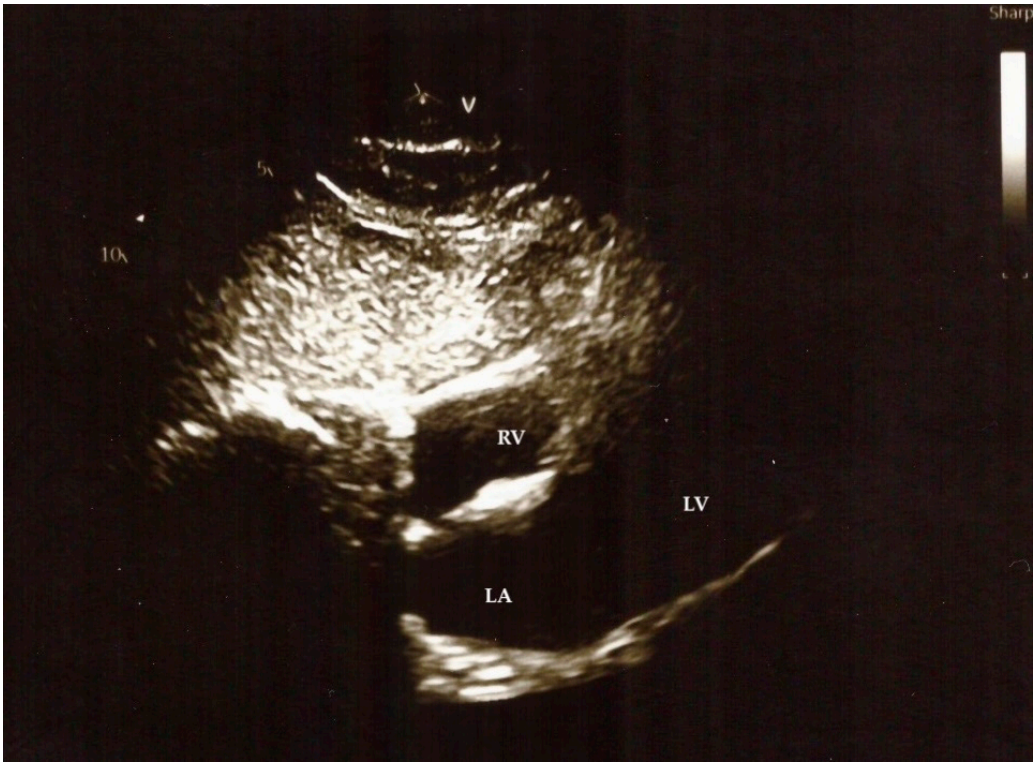


**Figure 2c.** TTE of Clinical Case 1: The subcostal view provided satisfactory image quality and most closely approximated standard imaging planes; LA – left atrium; LV – left ventricle; RV – right ventricle.

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The abnormal cardiac orientation on TTE, together with its limited diagnostic yield and the persistence of symptoms, raised suspicion for an underlying structural or positional abnormality and prompted further evaluation CMR, which ultimately established the diagnosis. It revealed complete congenital absence of the left pericardium, pronounced cardiac levoposition and clockwise rotation of the heart, with posterolateral displacement of the apex and approximately two-thirds of the atrial mass located within the left hemithorax. A key diagnostic feature was the interposition of lung parenchyma between the ascending aorta and the pulmonary artery, forming the characteristic "lingular sign". Additionally, there was extension of epicardial fat into the mediastinum (Figures 3 and 4).

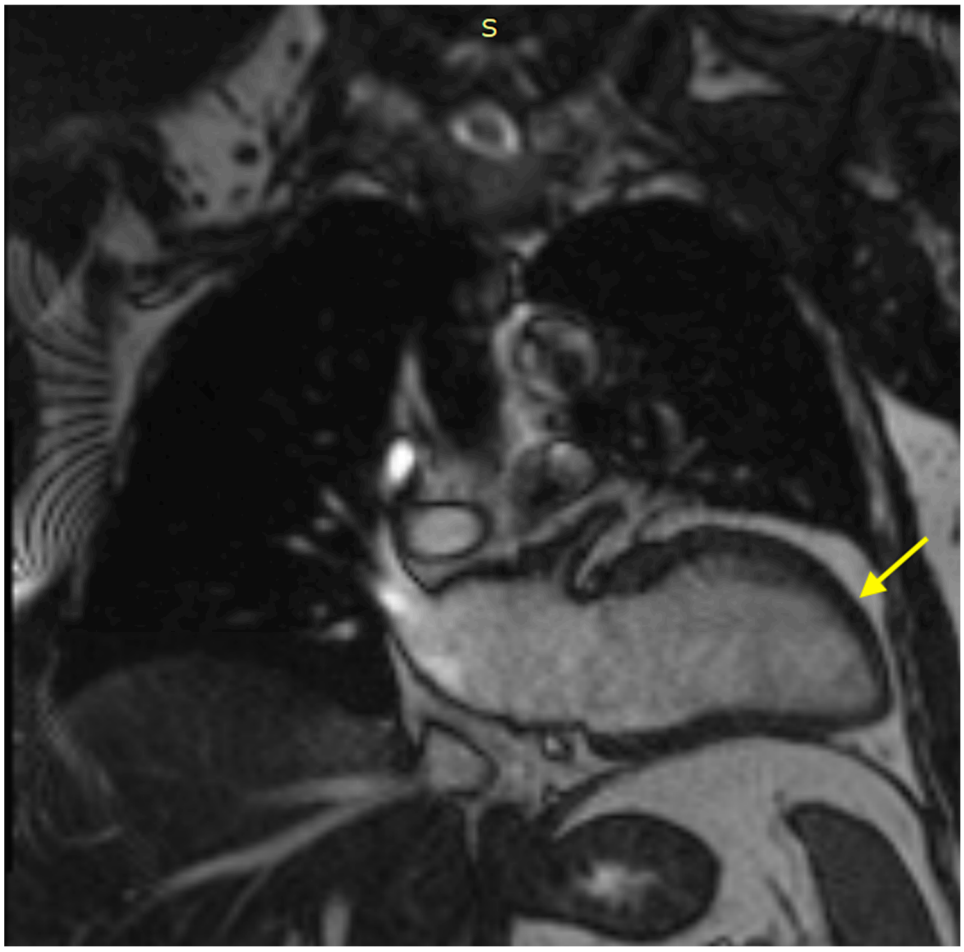


**Figure 3.** Cardiac MRI in the coronal plane with a large field of view, demonstrating abnormal leftward displacement of the cardiac silhouette within the left hemithorax (yellow arrow). .

Cardiac morphology and function were otherwise preserved, with normal biventricular volumes and systolic function and no evidence of myocardial edema or fibrosis. Incidental findings included mild mitral valve prolapse without significant regurgitation and a bovine aortic arch variant with a common origin of the brachiocephalic trunk and left common carotid artery. No pleural effusion, lymphadenopathy, or pulmonary pathology was identified.

Based on the absence of high-risk features and preserved cardiac function, a conservative management strategy was adopted. The patient continued his baseline medical therapy and was enrolled in a structured follow-up program, including clinical evaluation every six months, periodic laboratory monitoring, and annual electrocardiography and transthoracic echocardiography to assess cardiac position and exclude potential complications such as herniation.

This case highlights the diagnostic value of CMR in identifying congenital pericardial agenesis, particularly in patients with nonspecific symptoms, and underscores the importance of differentiating this entity from other structural or functional cardiac conditions.

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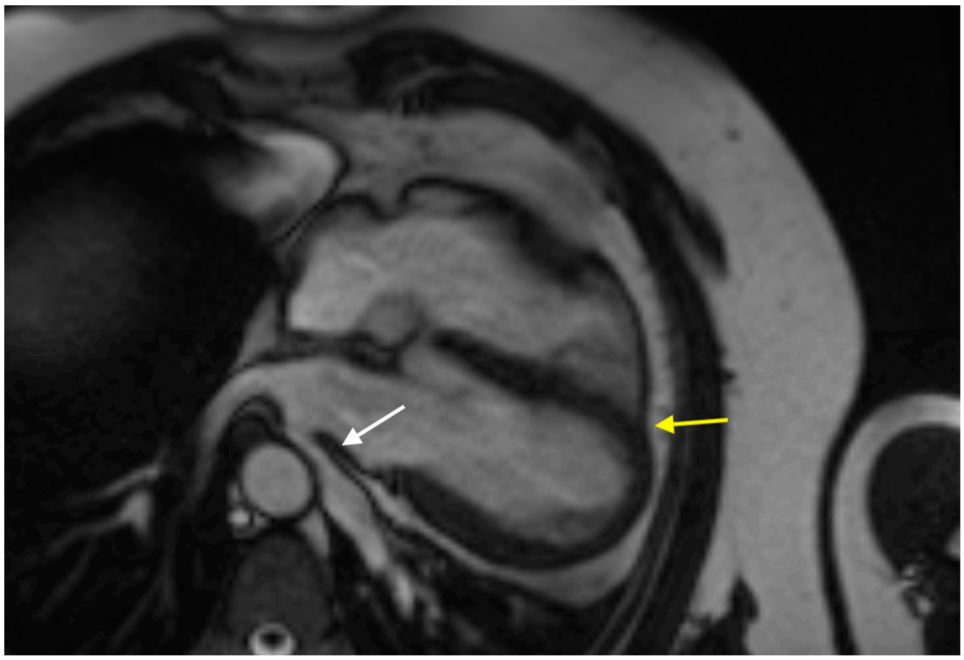

**Figure 4.** Cardiac magnetic resonance imaging (MRI) in the four-chamber cine view, confirming abnormal cardiac position with pronounced levorotation (yellow arrow). The inferior pulmonary vein is compressed between the descending aorta and the left atrium, likely as a consequence of the altered cardiac orientation (white arrow).

*Clinical case 2*

A 22-year-old male with a history of a cardiac murmur detected at birth, which had not been fully investigated, presented with intermittent chest discomfort described as stabbing pain in the precordial region. He had no history of syncope, palpitations, or exercise intolerance.

On physical examination, the patient was in good general condition, with a hypersthenic habitus and increased body mass. Vital signs were stable, with blood pressure of 100/70 mmHg. Cardiovascular examination revealed regular heart rhythm with clear heart sounds and a grade 2/6 systolic murmur best heard at the pulmonary area. No signs of heart failure were present. Laboratory investigations were within normal limits.

Electrocardiography demonstrated atrial rhythm with a heart rate of 84 bpm, a semi-horizontal electrical axis, and an incomplete right bundle branch block, considered a physiological finding for age.

Initial transthoracic echocardiography (TTE) raised suspicion for structural heart disease, suggesting right ventricular dilatation, possible secundum atrial septal defect, and the presence of pericardial effusion. These findings led to diagnostic uncertainty. Repeat TTE evaluation revealed abnormal cardiac orientation, with leftward rotation of the cardiac apex, apparent right ventricular enlargement, and signal drop-out in the region of the fossa ovalis without evidence of interatrial shunting.

To further clarify these findings, contrast-enhanced multislice cardiac CT with ECG synchronization was performed. CT imaging demonstrated significant lateral displacement (levoposition) and rotation of the heart, with the apex directed superiorly. There was absence of the pericardium along the left cardiac border, accompanied by interposition of lung parenchyma between the ascending aorta and the pulmonary artery, a characteristic diagnostic feature. In addition, coronal reconstructions showed an apparent “suspension” of the heart away from the diaphragm. Importantly, cardiac chambers, septa, valves, and coronary anatomy were normal, and no true pericardial effusion was identified (Figures 5 and 6).

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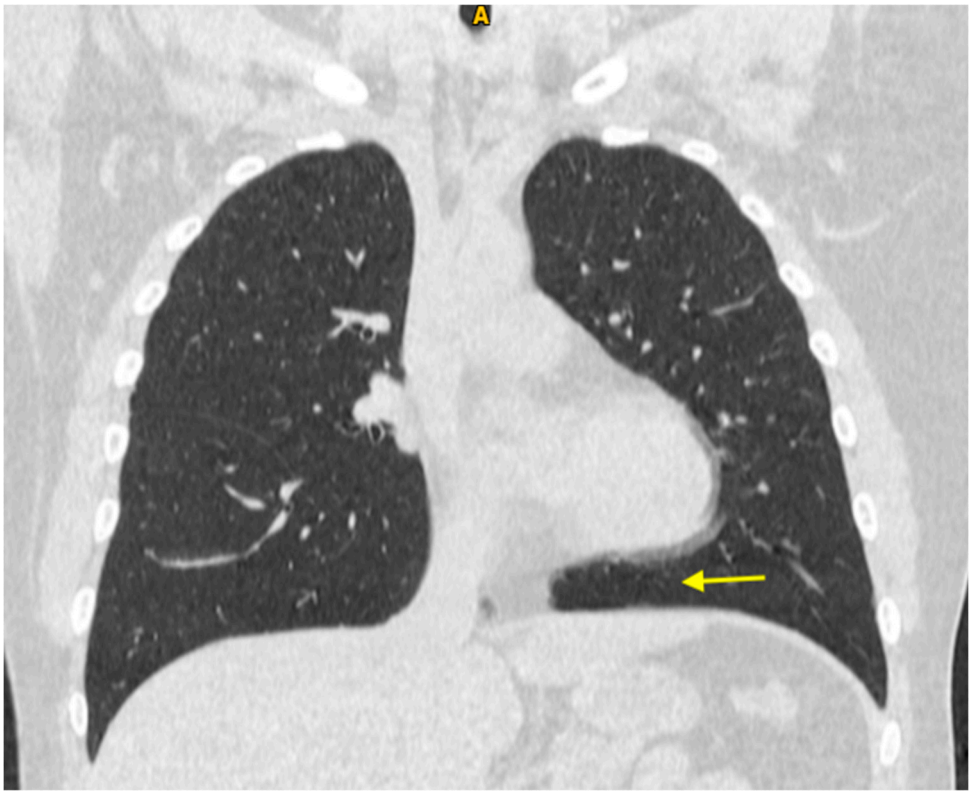


**Figure 5.** CT of the chest in the coronal plane using a lung window, demonstrating interposition of lung parenchyma between the left hemidiaphragm and the cardiac base (yellow arrow).

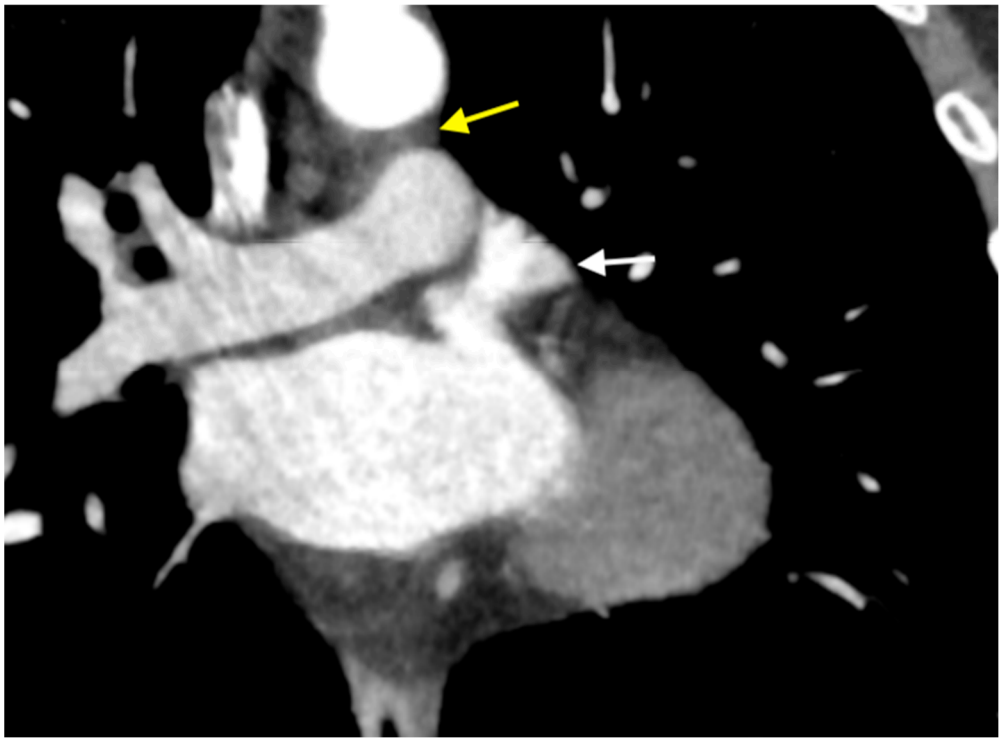

**Figure 6.** Contrast-enhanced CT of the heart in the coronal plane, demonstrating interposition of lung parenchyma between the aorta and the pulmonary artery (yellow arrow). A prominent left atrial appendage is also noted (white arrow).

These imaging findings established the diagnosis of congenital absence of the left pericardium.

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The patient remained hemodynamically stable and largely asymptomatic, apart from mild intermittent chest discomfort. Given the absence of high-risk features or complications, no specific cardiologic treatment was indicated. A conservative management strategy with regular clinical and imaging follow-up was recommended.

This case illustrates the potential for congenital pericardial agenesis to mimic structural heart disease, including atrial septal defect, right ventricular cardiomyopathy, and pericardial effusion on echocardiography, and highlights the essential role of advanced imaging in establishing the correct diagnosis.

*Clinical case 3*

A 29-year-old woman was initially evaluated in 2018 after cardiomegaly was incidentally identified during assessment for bronchopneumonia. At that time, TTE suggested dilatation of the right ventricular outflow tract (up to 42 mm) with suspected right ventricular systolic dysfunction, raising concern for possible arrhythmogenic right ventricular cardiomyopathy.

Subsequent echocardiographic examinations demonstrated persistent apparent enlargement of the right ventricular outflow tract (approximately 36 mm), with the cardiac apex appearing to be predominantly formed by the right ventricle. Left ventricular systolic function remained preserved (66%), and no significant valvular abnormalities were detected. Despite these findings, the clinical picture remained inconclusive.

In April 2023, the patient presented with an episode of panic attack accompanied by sinus tachycardia (heart rate 102 bpm) and mildly elevated blood pressure (130/80–85 mmHg), without chest pain or syncope. In 2024, she sought further medical evaluation following an episode of vertigo associated with transient blood pressure elevation.

On physical examination, the patient was in good general condition, with normal pulmonary findings. Cardiovascular examination revealed tachycardia and a grade 2/6 systolic murmur best heard at the second left intercostal space. Blood pressure was initially elevated (155/90 mmHg) but normalized spontaneously to 121/80 mmHg as heart rate decreased to 90 bpm.

Electrocardiography showed sinus tachycardia (123 bpm), a vertical electrical axis, and incomplete right bundle branch block. Twenty-four-hour Holter monitoring demonstrated sinus rhythm with rare supraventricular extrasystoles and no ventricular arrhythmias, pauses, or ischemic changes.

Given the persistent diagnostic uncertainty, CMR was performed. Imaging revealed complete congenital absence of the left pericardium, with pronounced leftward shift of the heart into the left hemithorax and clockwise rotation, resulting in leftward displacement of the apex. A key diagnostic feature was the interposition of lung parenchyma between the ascending aorta and the pulmonary artery. Despite the altered cardiac position, ventricular size, morphology, and systolic function were preserved, and no myocardial fibrosis or structural abnormalities were identified (Figures 7 and 8).

These findings established the diagnosis of complete left pericardial agenesis, explaining the apparent right ventricular enlargement observed on echocardiography as a positional artifact rather than true pathology.

Given the absence of symptoms attributable to structural heart disease and the lack of high-risk features, no specific treatment was required. The patient was managed conservatively with periodic clinical and imaging follow-up.

This case highlights the potential for congenital pericardial agenesis to mimic right ventricular cardiomyopathy or other structural abnormalities on echocardiography, and emphasizes the pivotal role of CMR in establishing a definitive diagnosis. It also illustrates the typically benign clinical course of isolated complete pericardial agenesis.

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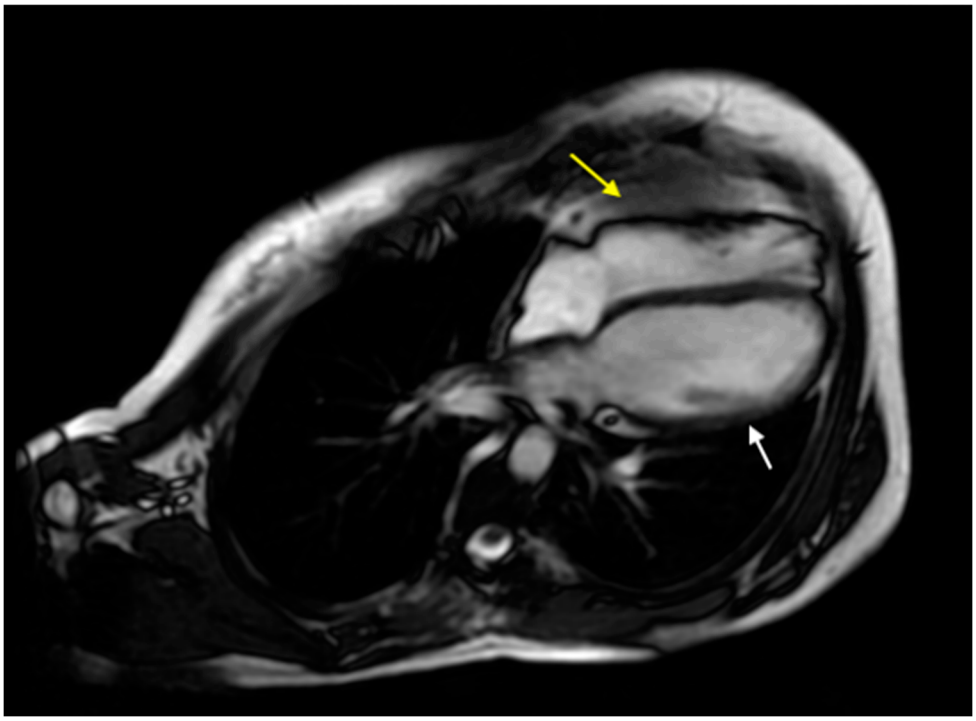

**Figure 7.** Cardiac MRI in the four-chamber cine view, demonstrating the pericardium as a thin linear structure anterior to the right ventricle, interposed between epicardial and mediastinal fat tissue (yellow arrow). The pericardium is not visualized posterior to the left-sided cardiac chambers (white arrow).

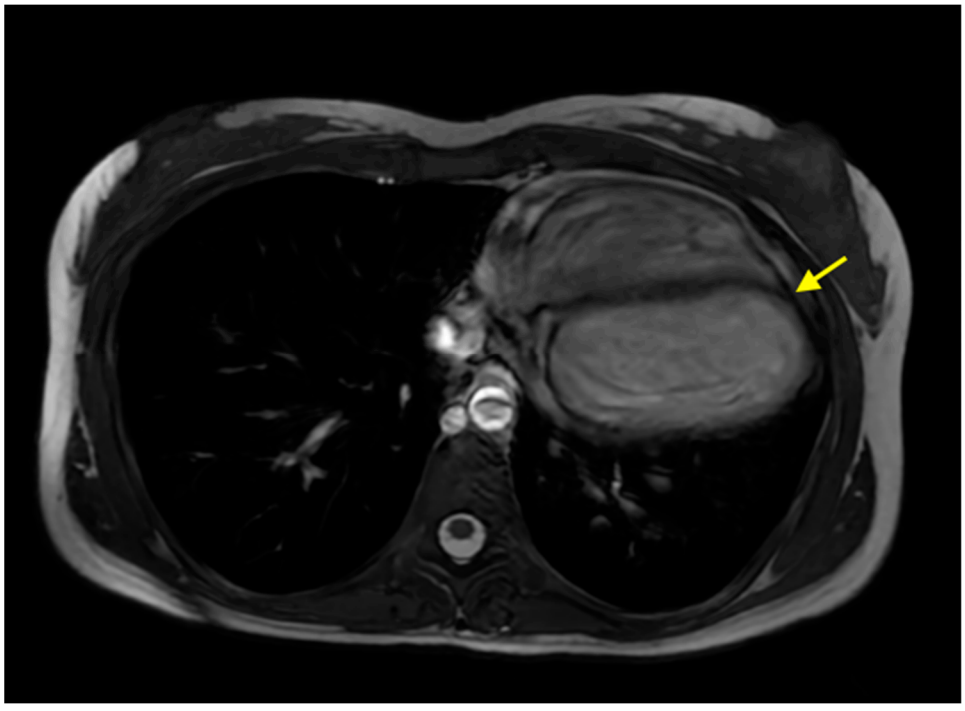

**Figure 8.** Cardiac MRI in the axial plane, demonstrating leftward displacement of the heart with rotation of the cardiac apex (yellow arrow).

*Clinical case 4*

A 62-year-old male patient was referred for cardiologic evaluation due to episodic, mild, transient chest oppression, predominantly left-sided, which had developed approximately two

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months prior to presentation. His medical history included arterial hypertension and mild dyslipidemia, both well controlled with medical therapy.

On physical examination, the patient was in good general condition. Vital signs were within normal limits (blood pressure 128/76 mmHg, heart rate 64 bpm). Cardiovascular and pulmonary examinations revealed no pathological findings. Laboratory investigations were unremarkable.

ECG demonstrated sinus rhythm at 67 bpm with left anterior fascicular block, without evidence of ischemic changes.

Initial TTE raised suspicion for pericardial pathology; however, image quality was limited due to a suboptimal acoustic window, precluding definitive assessment. In view of the persistent symptoms and inconclusive echocardiographic findings, the patient was referred for advanced imaging with CMR.

This investigation demonstrated partial congenital absence of the left pericardium. Imaging revealed marked leftward displacement and clockwise rotation of the heart, with the apex directed superiorly. Characteristic findings included interposition of lung parenchyma between the great vessels and beneath the heart, as well as prominent lobulated paracardiac fat along the left cardiac border. The right ventricle was positioned parallel to the sternum without evidence of dilatation.

Cardiac chambers, valvular structures, and ventricular systolic function were preserved, and no evidence of myocardial ischemia, fibrosis, or pericardial effusion was identified (Figures 9 and 10).

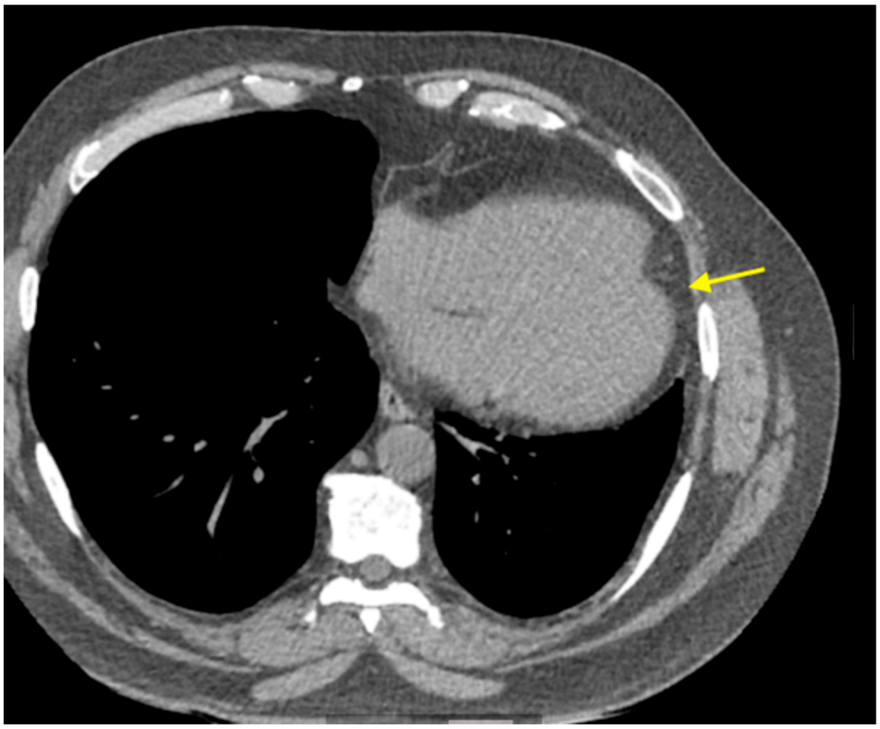

**Figure 9.** Non-contrast CT of the chest, demonstrating leftward displacement of the heart with associated rotation (yellow arrow).

Based on the absence of high-risk features such as herniation or myocardial compromise, a conservative management approach was adopted. The patient was advised to undergo regular clinical and imaging follow-up, with particular attention to the potential development of symptoms suggestive of mechanical complications.

This case is particularly relevant as it illustrates a symptomatic presentation of partial pericardial agenesis, a subtype associated with a higher risk of complications. It also highlights the diagnostic limitations of echocardiography in certain patients and underscores the critical role of advanced imaging modalities, particularly CMR, in establishing an accurate diagnosis.

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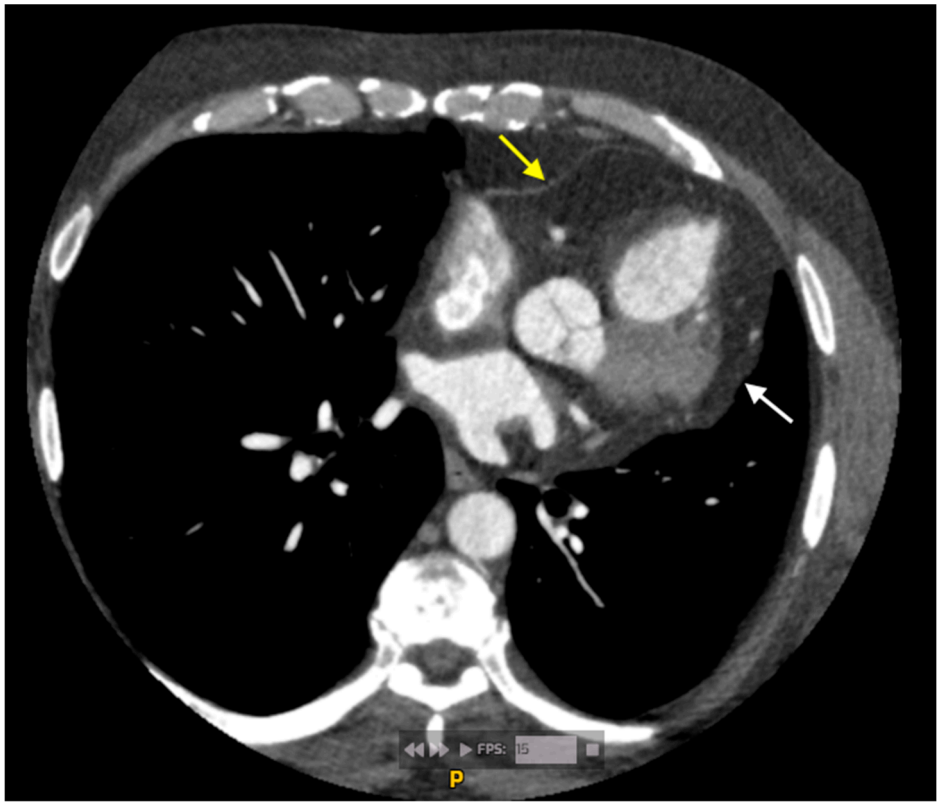


**Figure 10.** ECG-gated CT in the axial plane, demonstrating partial visualization of the pericardium anterior to the right-sided cardiac chambers (yellow arrow), with absence of the pericardium along the left side (white arrow).

*3.2. Summary of Findings*

Three of our patients had complete left pericardial agenesis, while one patient demonstrated a partial defect. Across all four cases, CPA was confirmed by multimodality imaging. The most consistent findings included:

- leftward displacement (levoposition) of the heart;
- clockwise cardiac rotation;
- interposition of lung parenchyma between mediastinal structures;
- absence of pericardial tissue, predominantly on the left side.

Clinical presentation was variable and nonspecific, ranging from asymptomatic cases to mild symptoms such as chest discomfort or dyspnea. Importantly, CPA mimicked other cardiac conditions on TTE in several cases, including cardiomyopathy, atrial septal defect, and pericardial effusion. ECG findings were nonspecific, typically showing sinus rhythm or tachycardia with axis deviation and minor conduction abnormalities (incomplete RBBB or left anterior fascicular block), without ischemic changes or significant arrhythmias.

None of the patients developed complications or required surgical intervention during the observation period.

Table 1 summarizes the clinical and imaging characteristics of the presented clinical cases.

**Table 1.** Summary of clinical and imaging characteristics.

| Case | Age/Sex | Presentation | Initial Suspicion | Imaging Modality | Type of CPA | Key Imaging Findings | Management |
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| | | | | | | | |
|---|---|---|---|---|---|---|---|
| 1 | 54/M | Dyspnea, fatigue | None | CMR | Complete | Levoposition, lung interposition, normal function | Conservative |
| 2 | 16/M | Chest pain | ASD, RV dilation, effusion | CT | Complete | Cardiac rotation, absent pericardium, lung interposition | Conservative |
| 3 | 29/F | Incidental cardiomegaly | RV cardiomyopathy | CMR | Complete | Left displacement, normal function | Conservative |
| 4 | 62/M | Incidental | None | CT | Partial | Lung interposition, partial defect | Conservative |

## 4. Discussion

Congenital pericardial agenesis is a rare developmental anomaly with a wide clinical spectrum, ranging from completely asymptomatic incidental findings to presentations mimicking structural or ischemic heart disease [5,12,18]. The present case series illustrates this variability and provides insight into the ongoing debate of whether CPA represents a benign anatomical variant or a clinically relevant condition.

In our cohort, all four patients demonstrated typical imaging features of left-sided pericardial absence, including cardiac levoposition, clockwise rotation, and interposition of lung parenchyma between mediastinal structures. Despite heterogeneous clinical presentations—ranging from exertional dyspnea and chest discomfort to incidental findings—none of the patients exhibited hemodynamic compromise or required surgical intervention. These observations are consistent with previous reports suggesting that complete pericardial agenesis is most often a benign condition, particularly when not associated with structural abnormalities [5,19,20].

The embryological basis of CPA is attributed to premature atrophy of the common cardinal veins (ducts of Cuvier), leading to defective formation and fusion of the pleuropericardial membranes during early gestation [2]. This results in absence of the pericardial sac and loss of its stabilizing function, allowing excessive cardiac mobility and displacement into the left hemithorax [12,14]. The predominance of left-sided defects is explained by the earlier regression of the left duct of Cuvier, impairing vascular supply to the developing pericardial membrane [5,17,21].

A key finding of our series is the high rate of initial misdiagnosis or diagnostic uncertainty, particularly on TTE. In two of our cases, CPA mimicked right ventricular enlargement, cardiomyopathy, or atrial septal defect, while in another case it raised suspicion for pericardial disease. These findings reflect the well-documented limitations of TTE in CPA, where altered cardiac position and acoustic windows may lead to misleading interpretations. Similar diagnostic pitfalls have been extensively described in the literature emphasizing that CPA should be considered in cases of unexplained right ventricular dilation or atypical cardiac orientation [5,22,23].

Multimodality imaging plays a central role in the diagnosis [5,20,24]. While chest radiography and ECG findings are nonspecific, cardiac CT and especially CMR provide definitive anatomical characterization [5,20,24]. The most reliable imaging features include marked leftward displacement of the heart, absence of the pericardial lining, and interposition of lung tissue between the ascending aorta and pulmonary artery—a hallmark sign of CPA characterization [5,20,24]. In our series, CMR was particularly valuable in resolving diagnostic ambiguity and confirming normal cardiac morphology and function. These observations align with current evidence identifying CMR as the gold standard for non-invasive diagnosis [5,14,16].

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The clinical relevance of CPA largely depends on the type of defect [5,12,23]. Complete pericardial agenesis, as observed in three of our patients, is generally well tolerated and rarely associated with complications. In contrast, partial defects carry a significantly higher risk, due to the presence of residual pericardial rims that may entrap cardiac structures [5,11,23]. Complications such as cardiac herniation, strangulation of the left atrial appendage or ventricular free wall, coronary artery compression, myocardial ischemia, and even sudden cardiac death have been reported [5,7,23].

Our fourth case is particularly illustrative in this regard. The patient presented with chest discomfort and was found to have a partial pericardial defect. Although no complications were observed, the symptomatic presentation highlights that CPA cannot always be considered an innocent finding, especially in the presence of partial defects. This reinforces the need for careful evaluation and follow-up in such patients.

Management strategies remain individualized due to the rarity of the condition and lack of large-scale studies [5,14,23]. Asymptomatic patients with complete pericardial agenesis generally require no specific treatment beyond periodic monitoring [5,14,23]. In contrast, symptomatic patients or those with partial defects may require closer surveillance or surgical intervention. Surgical options, including pericardioplasty or defect closure, are typically reserved for patients with significant symptoms or evidence of mechanical complications [5,7,14,23].

An important clinical implication of our study is that CPA should be actively considered in the differential diagnosis of unexplained cardiac displacement, suspected right ventricular pathology, or inconclusive echocardiographic findings. Failure to recognize this entity may lead to unnecessary diagnostic procedures, misdiagnosis, or inappropriate treatment.

Taken together, our findings support the concept that CPA represents a spectrum rather than a uniform entity. While complete forms are predominantly benign, partial defects may carry clinically relevant risks. Therefore, the question posed in the title—whether CPA is an innocent finding or a real clinical problem—does not have a binary answer. Instead, the clinical significance depends on the anatomical subtype, symptomatology, and presence of complications. The key imaging characteristics that support the diagnosis of CPA across modalities are summarized in Table 2.

**Table 2.** Key imaging features of congenital pericardial agenesis.

| Imaging Modality | Key Findings in CPA | Diagnostic Value | Limitations |
|---|---|---|---|
| **Chest X-ray** | Leftward cardiac displacement, elongated left heart border (“Snoopy sign”). | Initial suspicion | Nonspecific |
| **ECG** | Axis deviation, incomplete RBBB, poor R-wave progression. | Supportive only | Low specificity |
| **Echocardiography** | Apparent RV enlargement, abnormal cardiac orientation, unusual acoustic windows. | Raises suspicion | Frequently misleading |
| **Cardiac CT** | Absence of pericardium, lung interposition, cardiac levoposition, anatomy of coronaries. | High spatial resolution | Radiation exposure |
| **CMR (Gold standard)** | Direct visualization of absent pericardium, lung interposition, cardiac mobility, preserved function. | Definitive diagnosis | Limited availability |

## 5. Conclusions

Congenital pericardial agenesis is a rare and often underrecognized condition that may present with nonspecific symptoms or be detected incidentally during imaging. Our case series highlights the broad clinical spectrum of this anomaly, ranging from asymptomatic findings to presentations mimicking structural heart disease.

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Multimodality imaging, particularly CMR, plays a pivotal role in establishing a definitive diagnosis and avoiding misinterpretation of echocardiographic findings. The distinction between complete and partial pericardial defects is essential, as it directly influences clinical relevance and management. While complete pericardial agenesis is generally benign and can be managed conservatively, partial defects may be associated with an increased risk of complications and require closer surveillance. Therefore, congenital pericardial agenesis should not be universally regarded as an innocent finding, but rather as a condition requiring individualized assessment based on anatomical and clinical characteristics. Increased awareness among clinicians is crucial to ensure accurate diagnosis, appropriate follow-up, and avoidance of unnecessary interventions.

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## Abbreviations

The following abbreviations are used in this manuscript:

| | |
|---|---|
| Ao | Aortic |
| CMR | Cardiac magnetic resonance |
| CPA | Congenital pericardial agenesis |
| CT | Computed tomography |
| ECG | Electrocardiography |
| LA | Left atrium |
| LV | Left ventricle |
| RA | Right atrium |
| RV | Right ventricle |
| TTE | Transthoracic echocardiography |

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