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CASE REPORT

CLINICAL CASE SERIES

Isolated Apical Hypoplasia of the Left and Right Ventricle

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ABSTRACT

Isolated apical ventricular hypoplasia is an extremely rare congenital heart disease. We describe 2 cases, each affecting a different side, presenting with unique clinical and imaging characteristics not hitherto delineated in the literature. (J Am Coll Cardiol Case Rep 2024;29:102362) © 2024 The Authors. Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

CASE PRESENTATION

CASE 1. A 23-year-old woman presented to our center with dizziness and an exertion-related syncopal episode. She had a history of isolated left ventricular apical hypoplasia (ILVAH) diagnosed at 12 years of age and paroxysmal atrial fibrillation (Supplemental Figure 1). Four years previously, she had undergone successful cryoablation of the pulmonary veins. On admission, vital signs were within normal limits, apart from oxygen saturation of 88% on room air. Physical examination revealed marked splitting of the second heart sound.

Twelve-lead electrocardiography (**Figure 1**) showed a rhythm originating from the left anterior fascicle.

LEARNING OBJECTIVES

- To differentiate between the clinical manifestations of apical hypoplasia in the LV and RV.
- To deepen comprehension of the structural and hemodynamic changes linked to apical hypoplasia in the LV and RV.

The most recent transthoracic echocardiogram (**Figures 2A to 2C**, Videos 1 to 5) indicated absence of the left ventricular (LV) apex, biventricular systolic dysfunction, an LV ejection fraction of 42%, LV global longitudinal strain of −11.9% (**Figures 2D to 2F**, Video 6), restrictive-type diastolic dysfunction, a left atrial volume index of 140 mL/m^2, mild mitral and tricuspid regurgitation, a hypertrophic right ventricle (RV), and a high probability of pulmonary hypertension (pulmonary artery systolic pressure 144 mm Hg, mean pulmonary artery pressure 101 mm Hg). Cardiac magnetic resonance (CMR) confirmed the absence of the LV apex, along with the spherical morphology of the LV and a wrap-around and elongated RV (**Figure 3**, Video 7).

Pulmonary hypertension was confirmed through right heart catheterization (pulmonary capillary wedge pressure 9 mm Hg, mean pulmonary artery pressure 80 mm Hg, cardiac index 2.3), and a vasodilator test showed no significant response to inhaled iloprost. Following the exclusion of potential secondary causes of pulmonary hypertension, the diagnosis of class 1 pulmonary arterial hypertension was established. We initiated treatment by administering

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ABBREVIATIONS AND ACRONYMS

3D = 3-dimensional

ARVC = arrhythmogenic right ventricular cardiomyopathy

CMR = cardiac magnetic resonance

EAM = electroanatomical voltage mapping

ICD = implantable cardioverter-defibrillator

ILVAH = isolated left ventricular apical hypoplasia

LV = left ventricle/ventricular

RV = right ventricle/ventricular

TTE = transthoracic echocardiography

sildenafil (20 mg orally 3 times a day) and macitentan 10 mg per day. Because of the rhythm disorder and systolic dysfunction of the LV, physiological cardiac pacing was recommended by the cardiac team.

CASE 2. A 10-year-old child presented with syncope preceded by palpitations, occurring while standing and walking. His older brother had a history of sudden cardiac death at 18 years of age. Initial electrocardiography showed sinus rhythm, right bundle branch block, and left posterior fascicular block (Figure 4). On transthoracic echocardiography (TTE), the RV exhibited generalized hypokinesia, absence of the apex, thin and hyperreflective walls, and an aneurysmal interventricular septum with paradoxical septal motion, shifting toward the LV in systole. CMR revealed a spherical RV with the absence of its apical portion, an aneurysm in the basal and mid septum, dyskinesia of the free wall of the RV, fibrofatty infiltration of the RV free wall, an RV ejection fraction of 43%, and an LV ejection fraction of 49% (Figure 5, Table 1).

Implantation of an implantable cardioverter-defibrillator (ICD) was indicated because of syncope of unknown origin and the presence of arrhythmogenic RV cardiomyopathy (ARVC) (Class 2a, Level of Evidence: C)[1]; subcutaneous ICD implantation was ruled out because of T-wave sensing. Three-dimensional (3D) electroanatomical voltage mapping (EAM) was performed using the CARTO 3 System (Biosense Webster) to determine the appropriate site for endocardial implantation of the coil electrode. Voltage mapping was conducted using a multipolar catheter (PENTARAY NAV, Biosense Webster). Tissue with voltage < 0.5 mV was considered inactive or scar tissue; during the reconstruction of the RV, a scar was identified in the anterior wall and outflow tract, while the remaining RV had normal voltage (Figures 6A and 6B). The presence of this inactive zone is consistent with findings typically observed in patients with ARVC, as reported in other studies.[2]

Unipolar mapping (with a lower threshold of 5.2 mV) revealed a scar in the same epicardial area. An ICD (St. Jude Medical) was endocardially implanted, anchored in the right atrial appendage, and the high-power lead (single coil) was secured in the lowest and posterior part of the RV, where healthy tissue was present. The implantation parameters were within normal ranges, with ventricular sensitivity of 16.5 mV, impedance of 578 Ω, a stimulation threshold

FIGURE 1 12-Lead Electrocardiogram of Case 1

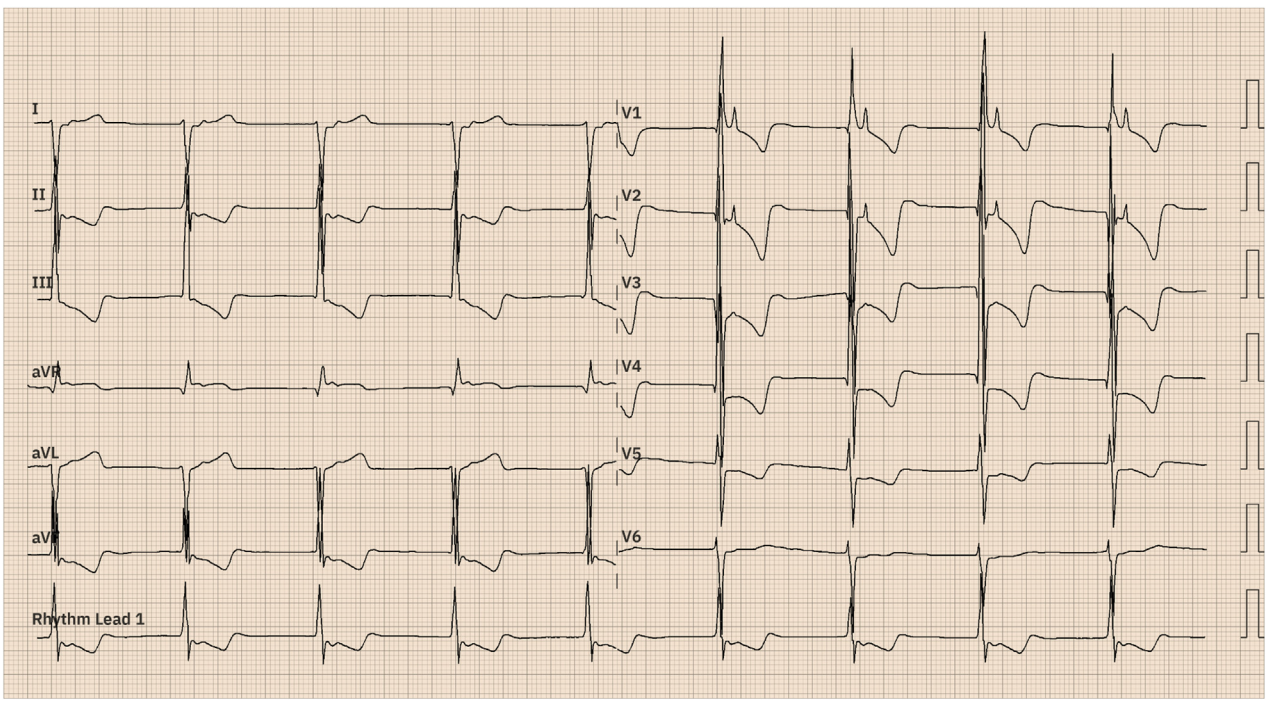
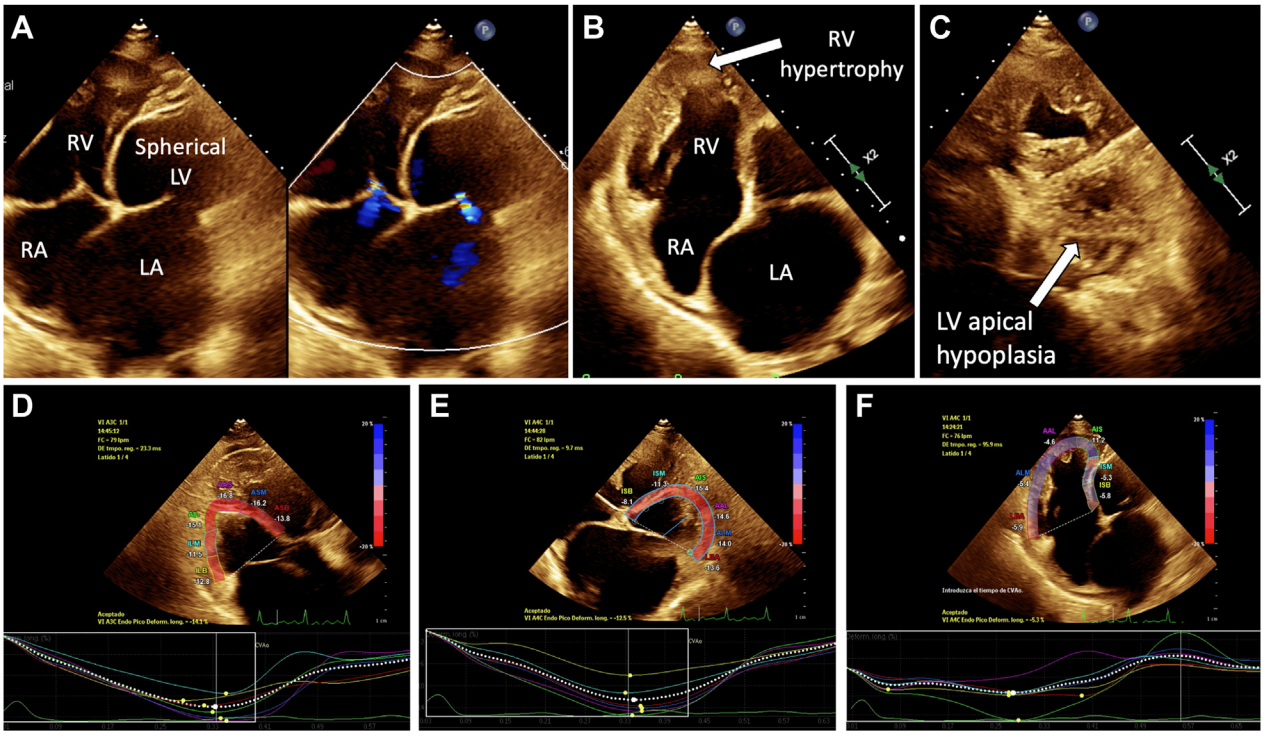
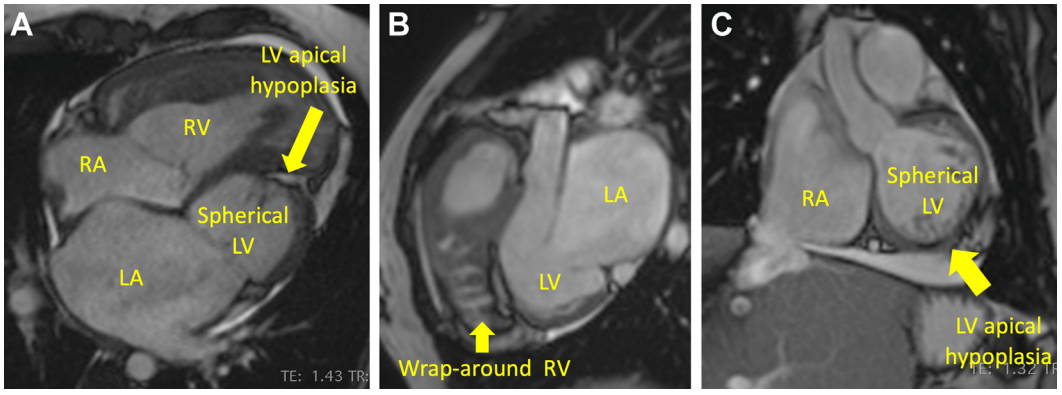


FIGURE 2 **Characterization by Transthoracic Echocardiography of Isolated LV Apical Hypoplasia**



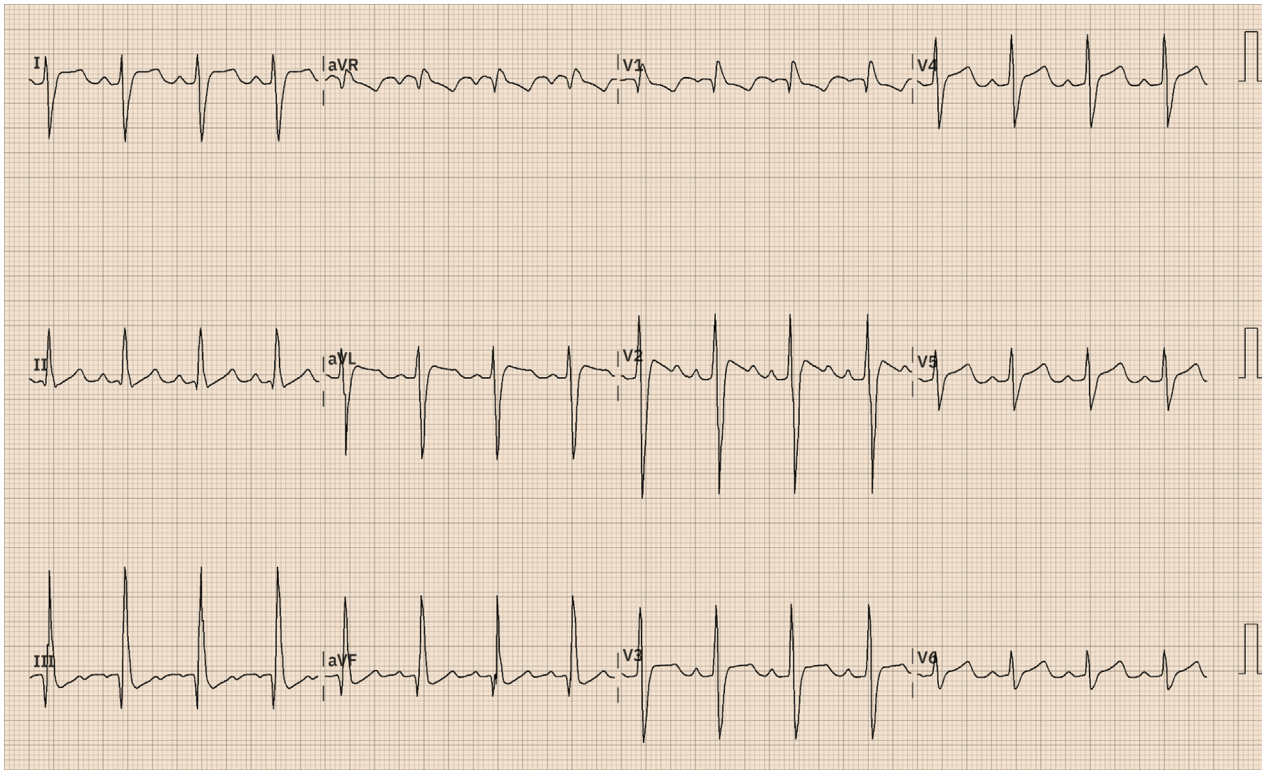
Apical 4-chamber view with color Doppler reveals a spherical left ventricle (LV) (A), and in another apical 4-chamber view (B), a hypertrophic right ventricle (RV) is observed, encompassing part of the LV apex. The left atrium (LA) appears severely dilated, and the interatrial septum is displaced to the right. In the parasternal short-axis view at the apex, there is apical hypoplasia of the LV with hyperreflectivity of the septum (C). (D to F) Analysis of LV and RV deformation in isolated LV apical hypoplasia. RA = right atrium.

FIGURE 3 **Characterization by Cardiac Magnetic Resonance of Isolated LV Apical Hypoplasia**



T1-weighted transverse long-axis 4-chamber view (A), sagittal long-axis 3-chamber view (B), and coronal view (C) highlighting the spherical LV as well as a wrap-around and elongated RV. Abbreviations as in Figure 2.

FIGURE 4 12-Lead Electrocardiogram of Case 2



of 0.4 mV, and a pulse width of 0.4 ms. Ventricular tachycardia and ventricular fibrillation zones were programmed at 170 and 222 beats/min, respectively. During a 60-month follow-up, 2 episodes of ventricular tachycardia were recorded, one managed with antitachycardia pacing and the second with a 40-J discharge.

Clinical, electrocardiographic, and imaging characteristics of the patients are summarized in **Table 2**.

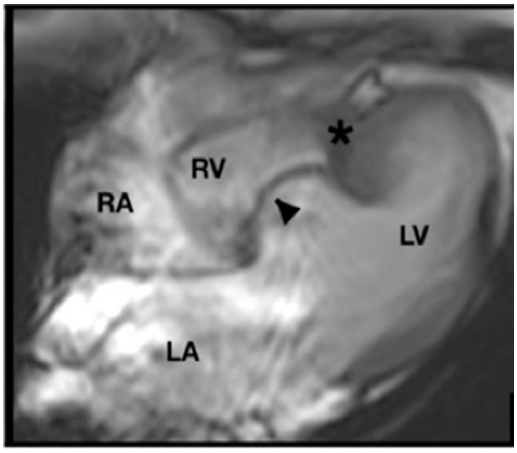
DISCUSSION

Ventricular hypoplasia is an exceedingly rare congenital myocardial disease characterized by varying degrees of underdevelopment of a ventricle and its components.[3,4] In cases of mild LV hypoplasia, patients may be asymptomatic or report mild dyspnea in adulthood, and secondary mitral regurgitation may also coexist.[5] In more severe cases, patients present from birth with pulmonary venous congestion, cyanosis, and tachypnea.[6] In a systematic review involving 37 cases of ILVAH, the main reported electrocardiographic abnormalities were T-wave abnormalities (29.7%), right-axis deviation with poor R-wave progression (24.3%), atrial fibrillation or flutter (24.3%), and nonsustained ventricular tachycardia (8.1%).[7]

Multimodal imaging is essential for diagnosis, as it provides crucial information for evaluating ventricular hemodynamic status and morphology and planning a therapeutic strategy.[3,5] Specific characteristics of ILVAH have been identified through CMR and TTE: a truncated and spherical LV and rightward bulging of the interventricular septum in diastole (CMR and TTE), a defective LV apex with infiltration of adipose tissue (CMR), a highly echogenic and akinetic apex (TTE), an abnormal origin of papillary muscles in the flattened LV apex (CMR and TTE), mild to moderate decreased contractility with a restrictive filling pattern (TTE), and an elongated, normally functioning RV wrapping around the deficient LV apex (CMR and TTE).[7,8]

Hypoplasia of the RV typically manifests with more severe symptoms, deeper cyanosis at birth, and higher mortality rates.[3] A systematic review and pooled analysis indicated that cyanosis was the most

FIGURE 5 Characterization by Cardiac Magnetic Resonance of Isolated RV Hypoplasia Associated With Arrhythmogenic RV Cardiomyopathy



Four-chamber bright-blood images reveal the absence of the apical portion of the RV (asterisk), an aneurysm of the interventricular septum with bidirectional displacement (arrow), irregularities in the RV wall in the basal and mid thirds due to fibroadipose infiltration, and a muscular band adjacent to the free wall of the RV. The tricuspid valve leaflet insertion is normal. Abbreviations as in Figure 2.

TABLE 1 European Task Force Criteria for the Diagnosis of Arrhythmogenic Cardiomyopathy

Category	RV Phenotype
I. Morphofunctional ventricular abnormalities	Major: aneurysm of the interventricular septum and systolic dysfunction of the right ventricle (RVEF 43%) Minor: dyskinesia of the free wall of the RV
II. Structural alterations	LGE in the free wall of the RV and the aneurysm of the interventricular septum
III. Repolarization abnormalities	No
IV. Depolarization and conduction abnormalities	No
V. Arrhythmias	Minor: 2 episodes of VT during the postimplantation clinical follow-up of the ICD, with one successfully managed using ATP and the second requiring a 40-J discharge
VI. Family history/genetics	Minor: premature sudden death (age <35 y) due to suspected sudden cardiac arrest in a first-degree relative
Diagnostic criteria	Meets diagnostic criteria for ARVC with 1 major criterion and 4 minor criteria.

ARVC = arrhythmogenic right ventricular cardiomyopathy; ATP = antitachycardia pacing; ICD = implantable cardioverter-defibrillator; LGE = late gadolinium enhancement; RV = right ventricle/ventricular; RVEF = right ventricular ejection fraction; VT = ventricular tachycardia.

prevalent symptom (92.6%), followed by digital clubbing (35.2%), dyspnea (31.5%), heart failure (9.3%), and arrhythmias (5.6%).[3] Cyanosis is attributed primarily to the presence of a patent foramen ovale or an atrial septal defect in most cases.[3]

Multimodal imaging reveals the absence of the trabecular portion of the RV (CMR and TTE), the presence of normally developed tricuspid and pulmonary valves (CMR), a truncated RV with a bulging interventricular septum toward the LV (CMR and TTE), tricuspid papillary muscles with an anomalous origin from a flattened apex (CMR and TTE), a relatively elongated LV wrapping around the deficient RV apex (CMR and TTE), and fatty infiltration in the ventricular apex, particularly along the distal interventricular septum (CMR).[9]

The syncope of unknown origin and the presence of ARVC prompted us to opt for the implantation of an ICD guided by 3D EAM, aiming to steer clear of ventricular lead placement in areas with low voltage. This choice was driven by evidence of complications with the devices in as many as 24% of patients with ARVC during postimplantation follow-up.[10] Furthermore, long-term observations have indicated a notable decline in ventricular sensitivity.[10]

CONCLUSIONS

ILVAH and isolated RV apical hypoplasia are 2 exceptionally rare cardiac malformations. This represents the inaugural documentation of 2 cases that juxtapose the clinical and imaging presentations not hitherto delineated in the literature. The initial case was a patient with ILVAH and severe class I pulmonary arterial hypertension, while the second involved a pediatric patient with isolated RV apical hypoplasia and ARVC (this is the sole documented case of the concurrent existence of these entities). This scenario raises distinctive therapeutic considerations, notably the implantation of an ICD prompted by syncope of unknown origin and ARVC, with guidance on 3D EAM. Using multimodal imaging is pivotal to attain the morphofunctional diagnosis of these 2 congenital anomalies and to formulate an effective therapeutic strategy.

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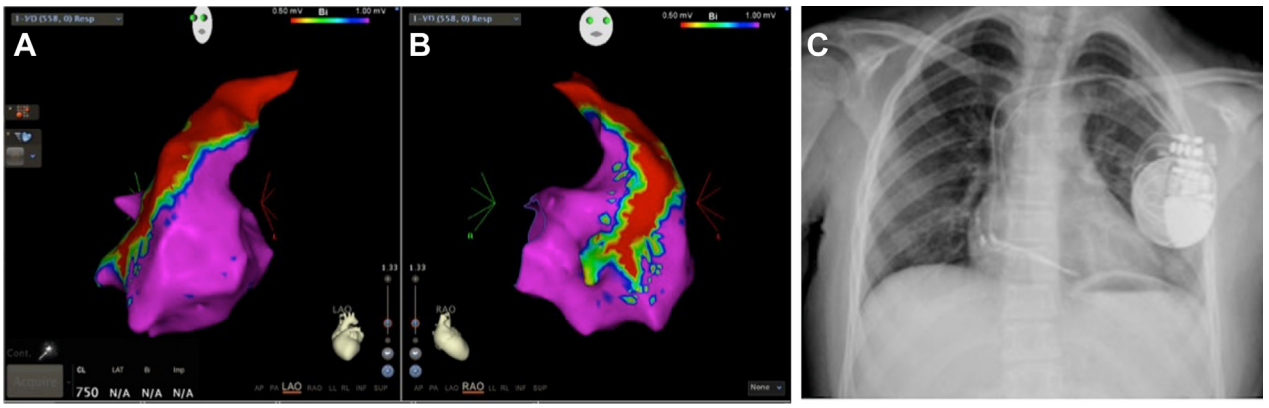
FIGURE 6 3-Dimensional Electroanatomical Voltage Mapping of the RV and Chest Radiography After Implantable Cardioverter-Defibrillator Insertion



CARTO mapping in the left anterior oblique (LAO) projection (A) of the right ventricular (RV) model shows an extensive electroanatomical scar in the anterior wall of the RV (red indicates voltage <0.5 mV, considered inactive or scar tissue), while in the right anterior oblique (RAO) projection (B), an electroanatomical scar is identified in the RV outflow tract. (C) Chest radiography after implantable cardioverter-defibrillator insertion.

TABLE 2 Clinical, Electrocardiographic, and Imaging Features

	Case 1	Case 2
Sex, age	Female, 23 y	Male, 10 y
Clinical presentation	Dizziness, exertional syncope	Palpitations, syncope
Arrhythmia	AF	No arrhythmia was identified
ECG	Rhythm originating from the left anterior fascicle with a frequency of 54 beats/min	Sinus rhythm, RBBB, and LPFB
Echocardiography	Agenesis of the LV apex, dilation of both right and left heart chambers, biventricular systolic dysfunction, LVEF 45%, LVGLS −11.9%, restrictive-type diastolic dysfunction, hypertrophic RV and PAH (PASP 144 mm Hg and mPAP 101 mm Hg)	Absence of RV apex, generalized hypokinesia, aneurysmal interventricular septum with paradoxical septal motion, with systolic shift toward the LV
MRI	Absence of the LV apex, along with spherical morphology of the LV and a wrap-around and elongated RV	Spherical RV with absence of its apical portion, dyskinesia and fibrosis of the free wall of the RV, aneurysm of the basal and middle third of the septum, RVEF 43%, LVEF 53%, LGE in the free wall of the RV and the aneurysm of the interventricular septum
Management	Cryoablation of pulmonary veins and initial dual oral combination therapy for PAH	ICD implantation guided by 3D EAM
Follow-up	Patient was scheduled for physiological cardiac pacing, followed by initiation of phase II cardiac rehabilitation; NYHA functional class II	During 60-mo follow-up, two episodes of VT were recorded, one managed with ATP and the second with a 40-J discharge; NYHA functional class I

3D = 3-dimensional; AF = atrial fibrillation; EAM = electroanatomic voltage mapping; ECG = electrocardiography; LPFB = left posterior fascicular block; LV = left ventricle/ventricular; LVEF = left ventricular ejection fraction; LVGLS = left ventricular global longitudinal strain; mPAP = mean pulmonary artery pressure; MRI = magnetic resonance imaging; PAH = pulmonary arterial hypertension; PASP = pulmonary artery systolic pressure; RBBB = right bundle branch block; other abbreviations as in Table 1.

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KEY WORDS **arrhythmogenic right ventricular cardiomyopathy, congenital heart disease, multimodal imaging, ventricular hypoplasia**

APPENDIX **For a supplemental figure and videos, please see the online version of this paper.**