ESC European Society of Cardiology

European Heart Journal - Case Reports (2023) **7**, 1–8
https://doi.org/10.1093/ehjcr/ytad046

CASE REPORT
Cardiovascular imaging

Isolated left ventricular apical hypoplasia: case report

Ricardo Román [1]*, Henry Anchante [1], Katia Menacho [2], and Félix Medina [1]

[1]Division of Cardiology, Cayetano Heredia Hospital, San Martin de Porres, Lima 15102, Perú; and [2]Barts Heart Centre, St Bartholomew's Hospital, W Smithfield, London EC1A 7BE, UK

Received 5 March 2022; first decision 8 April 2022; accepted 25 January 2023; online publish-ahead-of-print 7 February 2023

Background Isolated left ventricular apical hypoplasia (ILVAH) is a rare, probably congenital, cardiac condition recently reported in the last two decades. Although most cases are asymptomatic or mildly symptomatic, some severe and fatal cases have been reported, leading to more efforts for appropriate diagnosis and treatment. We describe the first, and severe, case of this pathology in Peru and Latin America.

Case summary A 24-year-old male with a history of long-standing alcohol and illicit drug use presented with symptoms of heart failure (HF) and atrial fibrillation (AF). Transthoracic echocardiography showed biventricular dysfunction along with spherical left ventricle, abnormal papillary muscles' origin from the left ventricular apex, and an elongated right ventricle wrapping around the deficient left ventricular apex. Cardiac magnetic resonance confirmed these findings and revealed subepicardial fatty replacement at the left ventricular apex. The diagnosis of ILVAH was made. He was discharged from hospital with carvedilol, enalapril, digoxin, and warfarin. Eighteen months later he remains mildly symptomatic at New York Heart Association functional class II without worsening HF nor thrombo-embolism.

Discussion This case highlights the usefulness of multimodality non-invasive cardiovascular imaging for accurate diagnosis of ILVAH as well as the importance of close follow-up and treatment of established complications (HF and AF).

Keywords Isolated left ventricular apical hypoplasia • Heart failure • Atrial fibrillation • Echocardiography • Cardiac magnetic resonance • Case report

ESC Curriculum 2.2 Echocardiography • 2.3 Cardiac magnetic resonance • 9.7 Adult congenital heart disease

Learning points

- Well-established imaging criteria at echocardiography and cardiac magnetic resonance enable diagnosis of isolated left ventricular apical hypoplasia.
- Disease-specific therapy does not exist, then continuous follow-up and treatment of established complications are imperative.

Introduction

Isolated left ventricular apical hypoplasia (ILVAH) is a rare, probably congenital, cardiac condition described since 2004.[1] Case reports have increased through years with a wide clinical spectrum ranging from asymptomatic children[2] to severely symptomatic adults with associated heart failure (HF), pulmonary hypertension (PH), and malignant tachyarrhythmias.[3,4] Hence, we present the first case of ILVAH

* Corresponding author. Tel: +51 968 517 989, Fax: +51 511 482 1410, Email: ricardo.roman@upch.pe
Handling Editor: Suzan Hatipoglu
Peer-reviewers: A Shaheer Ahmed; Mohammed Al-Hijji; Andriana Anagnostopoulou
Compliance Editor: Megha Agarwal
Supplementary Material Editor: Tinka Julia van Trier

reported in Peru and Latin America of a young adult who developed some of the aforementioned complications.

Timeline

8 years before admission	A 24-year-old male patient started cannabis, alcohol, and cocaine consumption.
1 year before admission	Palpitations onset (once a week), concomitant with drug use.
2 months before admission	More frequent palpitations (twice or more a week), with and without drug use. Onset of exertional dyspnoea. Drug use was discontinued.
Day 0 (day of admission)	He attended the emergency room due to decompensated heart failure (HF) and atrial fibrillation (AF) with rapid ventricular response and received acute medical treatment. Anticoagulation and rate control therapy were started with warfarin and carvedilol plus digoxin, respectively.
Day 1	Transthoracic echocardiogram (TTE) showed biventricular systolic dysfunction along with spherical left ventricle, abnormal papillary muscles' origin from left ventricular (LV) apex and elongated right ventricle wrapping around the LV apex, suggesting the possible diagnosis of isolated left ventricular apical hypoplasia (ILVAH).
	Diagnosis of HF with reduced ejection fraction (HFrEF) was made and treated with enalapril in addition to carvedilol.
Day 5	Cardiac magnetic resonance displayed the same TTE findings and showed subepicardial LV apex fatty replacement with no evidence of myocardial fibrosis. Diagnosis of ILVAH was confirmed.
2 weeks after hospital discharge	Mildly symptomatic patient with New York Heart Association (NYHA) functional class II and AF with rapid ventricular response. Carvedilol was up-titrated. Other HFrEF medications were limited due to low-normal blood pressure. Adjustment of warfarin dose due to high international normalized ratio (INR).
18 months after hospital discharge	Persistent AF with controlled ventricular response. Still on NYHA functional class II. No episode of thrombo-embolism reported. New TTE with similar findings as those of first study.

Case presentation

A 24-year-old male attended the emergency department with a 2-day history of persistent palpitations and dyspnoea. He reported such symptoms over the last year, which became more frequent over the last 2 months. He had no previous medical history but alcohol and illegal drug use (including cannabis and cocaine), all of them discontinued due to symptoms' exacerbation. His family history was unremarkable.

On admission, he was haemodynamically stable and his blood pressure was 128/80 mmHg. He had irregular tachycardia and bilateral crackles consistent with acute heart failure (HF). Electrocardiogram (ECG) showed atrial fibrillation (AF) with rapid ventricular response and non-specific intraventricular conduction disturbance (*Figure 1*). Chest X-ray confirmed pulmonary congestion (*Figure 2*). Laboratory findings revealed normal thyroid profile. He was given intravenous furosemide 40 mg and deslanoside 0.4 mg with clinical improvement.

The following day, the transthoracic echocardiogram (TTE) revealed severe left ventricular (LV) systolic dysfunction with an ejection fraction of 17%, increased LV filling pressure with an E/e' ratio 15, mild mitral regurgitation and severe left atrium dilation with an indexed volume of 63.4 mL/m^2. Furthermore, dysfunction of the right ventricle (RV) with a tricuspid annular plane systolic excursion (TAPSE) of 10 mm was present. In addition, striking features were noted. Both papillary muscles originated from a flattened LV apex (*Figure 3A*, see Supplementary material online, *Video S1*) while a spherical left ventricle and an elongated RV wrapping around the deficient LV apex resembled an orange-shaped and banana-shaped ventricle, respectively (*Figure 3B*, see Supplementary material online, *Video S2*).

Isolated left ventricular apical hypoplasia was suggested as a possible diagnosis and the patient was referred for an advanced cardiac magnetic resonance (CMR) scan. Despite low-quality images due to AF and low patient compliance with respiratory instructions, CMR displayed same findings as TTE with the addition of subepicardial fatty replacement at the LV apex (*Figure 4*, see Supplementary material online, *Video S3*) confirming ILVAH diagnosis. There was no clear evidence of myocardial fibrosis at late gadolinium enhancement sequence (*Figure 5*).

Diagnosis of HF with reduced ejection fraction (HFrEF) and AF with an intermediate risk of stroke (CHA2DS2-VASc = 1) was made. Toxins (particularly alcohol and cocaine), tachycardia-induced cardiomyopathy (TCM) and ILVAH *per se* were considered as potential explanations of ventricular dysfunction. Psychiatry counselling was encouraged to maintain drug abstinence. Moreover, AF ablation was proposed either because of the possibility of TCM or the coexistence of LV systolic dysfunction (irrespectively of its cause). Unfortunately, procedure-related costs precluded its performance. Then, the patient was started on carvedilol 6.25 mg twice daily (BD) and enalapril 10 mg BD, along with digoxin 0.25 mg once daily (OD) for rate control therapy due to low-normal blood pressure. Given low bleeding risk (HASBLED = 0) and severe left atria dilation, warfarin 5 mg OD was added for anticoagulation. He was discharged without signs of pulmonary congestion.

At 2 weeks of follow-up visit, he was at New York Heart Association (NYHA) functional class II and reported frequent palpitations with evidence of AF with rapid ventricular response at new ECG. Heart failure medication was up-titrated and warfarin dose adjusted due to high international normalized ratio (INR).

At present, 18 months later, he remains on NYHA functional class II and persistent AF with controlled ventricular response, with no episode of thrombo-embolism or bleeding. New TTE showed similar findings as those of first study.

Discussion

Isolated left ventricular apical hypoplasia is considered a likely congenital cardiac disorder as it has been reported at all age groups from infants, even foetuses, to older adults.[2,5] No specific sex predilection has been noted. In addition, concomitant congenital cardiac lesions have been recognized such as patent ductus arteriosus,[6] aortic stenosis, infundibular pulmonary stenosis,[7] atrial septal defect[8]; neither of them presents in this patient. Pathogenesis of this entity is still unknown with impaired ventricular partition[1] and hypoplasia of the apical trabecular component[9] as potential explanations. In this regard, genetics' role is being

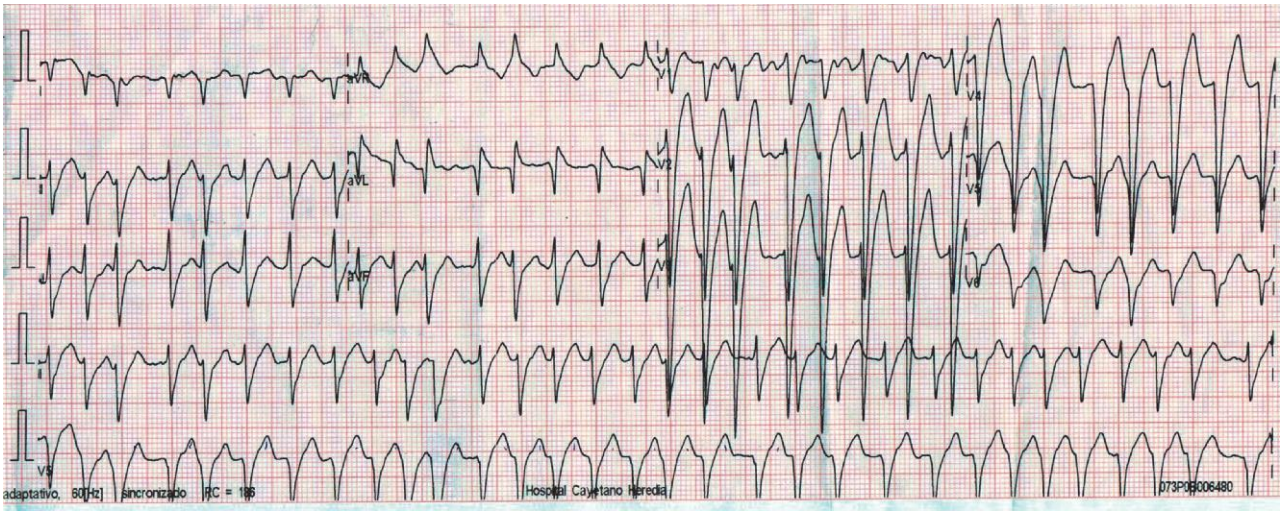

Figure 1 Electrocardiogram showing atrial fibrillation with rapid ventricular response and non-specific intraventricular conduction disturbance.

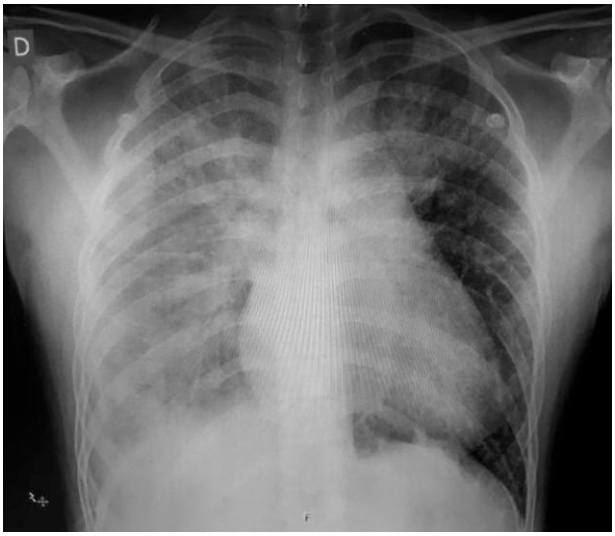

Figure 2 Chest X-ray showing pulmonary congestion.

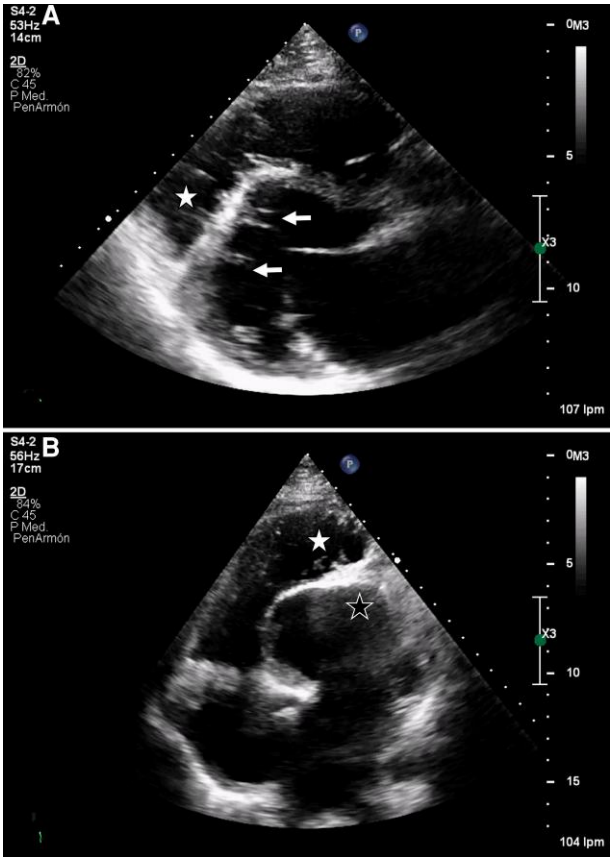

Figure 3 Transthoracic echocardiogram, 2D images on parasternal long-axis view (*A*) and apical four-chamber view (*B*) displaying isolated left ventricular apical hypoplasia features: spherical left ventricle (black star), abnormal papillary muscles' origin from the left ventricular apex (white arrows), and an elongated right ventricular wrapping around the deficient left ventricular apex (white star).

investigated[10] particularly after the association of this condition with a lamin A/C and NEXN gene mutations in previously reported cases.[11,12] Unfortunately, our patient did not have access to these genetic studies due to cost-related reasons. Family pattern has also been proposed, however, no single report has confirmed this hypothesis.

Clinical presentation is variable but mostly corresponding to mild cases (*Table 1*). Nevertheless, severe non-fatal[3] and fatal[4] cases have been reported as in this case with the development of HFrEF and AF. Physical exam is usually unremarkable but frequently showing a systolic murmur presumably due to coexistent congenital cardiac abnormality[6–8] or secondary mitral regurgitation.[1,4]

Electrocardiogram findings are often non-specific but displayed in some cases T-wave abnormality,[6] right axis deviation,[1,7] and intraventricular conduction disturbance,[5–8] which was noticed in our patient. An interesting association with Type 1 Brugada pattern has recently been

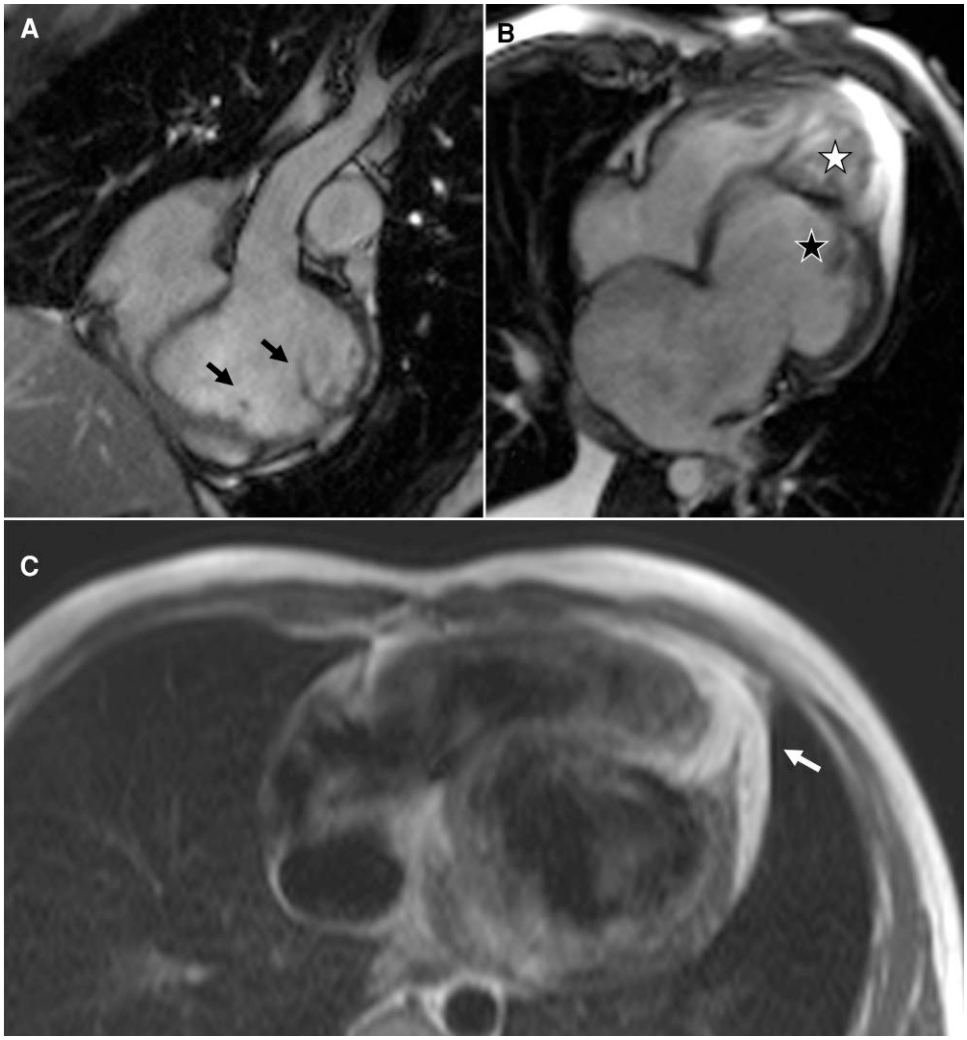

Figure 4 Cardiac magnetic resonance, left ventricular outflow tract view (*A*) and four-chamber view (*B*) showing spherical left ventricle (black star), abnormal papillary muscles' origin from the left ventricular apex (black arrows), and an elongated right ventricular wrapping around the deficient left ventricular apex (white star). Fat saturation image (*C*) displaying subepicardial fatty replacement at the left ventricular apex (white arrow).

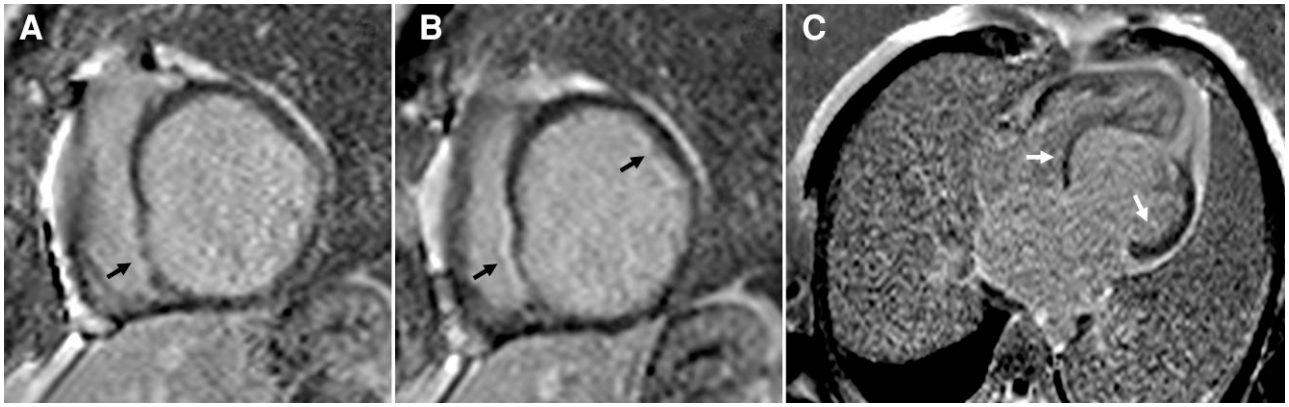

Figure 5 Cardiac magnetic resonance, base—mid short-axis view late gadolinium enhancement sequence (*A*, *B*) showing hyperenhancement at right ventricular side of inferior septum and basal anterolateral wall of left ventricle (black arrows) not seen at four-chamber view (*C*) late gadolinium enhancement sequence (white arrows). No clear evidence of myocardial fibrosis.

Table 1 Summary of clinical, electrocardiographic, and imaging features of left ventricular apical hypoplasia cases

Authors (year)	Sex and age	Clinical presentation	ECG	Multimodality imaging (TTE, CCT, and CMR)					Genetic mutation	Management	Follow-up	Outcomes
				Morphological features				Functional features				
				ILVAH findings[a]	FR	Associated congenital abnormalities	LGE					
Fernandez-Valls *et al.*[1] (2004)	F, 22 y	Mild dyspnoea, fatigue, or chest discomfort	SR, RAD, PRWP, low QRS voltage	Present	LV apex, subepicardial	NR	NR	Mild LV systolic dysfunction, normal RV systolic function, moderate MR, PH NR	NR	Standard HF treatment[b]	[b]	Improvement[b]
	F, 46 y		SR, RAD, PRWP, low QRS voltage	Present	LV apex, subepicardial	NR	NR	Mild-moderate LV systolic dysfunction, normal RV systolic function, MVP with MR severity NR, PH NR	NR	Standard HF treatment[b]	[b]	Improvement[b]
	M, 26 y		SR, RAD, PRWP, low QRS voltage	Present	LV apex, subepicardial	NR	NR	Moderate-severe LV systolic dysfunction, normal RV systolic function, no MR, PH NR	NR	Standard HF treatment[b]	[b]	Improvement[b]
Marin *et al.*[2] (2007)	M, 3 mo	Asymptomatic	NR	Present	NR	NR	NR	LV systolic function NR, normal RV systolic function, no MR, PH NR	NR	NR	NR	NR
Irving *et al.*[4] (2009)	M, 19 y	Cardiac arrest	AFl, VF, AF	Present	NR	NR	NR	Severe LV systolic dysfunction, RV systolic dysfunction, moderate MR, PH	NR	CICU admission (MV, inotropic support)	NR	In-hospital death
Motwani *et al.*[13] (2011)	M, 63 y	Dyspnoea, severity NR	AF	Present	LV apex, subepicardial	NR	Basal anteroseptal, mid-wall	Severe LV systolic dysfunction, RV systolic function NR, MR NR, PH NR	NR	Rate control therapy, then elective DC cardioversion	[b]	Symptoms improvement

Continued

Table 1 *Continued*

Authors (year)	Sex and age	Clinical presentation	ECG	Multimodality imaging (TTE, CCT, and CMR)					Genetic mutation	Management	Follow-up	Outcomes
				Morphological features				Functional features				
				ILVAH findings[a]	FR	Associated congenital abnormalities	LGE					
Haffajee *et al.*[6] (2011)	M, 50 y	Asymptomatic	NSICD, TWA	Present	LV apex, subepicardial	Ligated PDA	Absent	Moderate-severe LV systolic dysfunction, normal RV systolic function, no MR, no PH	NR	BB and ACEI	NR	NR
Vanhecke *et al.*[9] (2011)	F, 53 y	Palpitations, severity NR	SR, TWA, PRWP	Present	LV apex	NR	NR	Mild systolic dysfunction, RV systolic function NR, mild MR, PH NR	NR	Standard HF treatment[b]	NR	NR
Moon *et al.*[7] (2013)	M, 33 y	Asymptomatic	SR, RAD, incomplete RBBB, RVH, RAE	Present	LV apex	Infundibular PS, AS	Absent	Normal LV systolic function, normal RV systolic function, mild MR, PH NR	NR	Observation	[b]	[b]
Baroni *et al.*[8] (2014)	M, 45 y	Dyspnoea, severity NR	AF, complete LBBB	Present	LV apex	NR	Posterior insertion of the RV free wall in the VS, mid-wall	Moderate-severe LV systolic dysfunction, RV systolic dysfunction, mild MR, PH	NR	BB, ACEI and MRA. Denied ICD	02 y	Symptoms improvement, but moderate-severe systolic dysfunction persisted
	F, 12 y	Asymptomatic	NR	Present	Absent	No residual ostium primum ASD	Trabeculae of both ventricles	Mild systolic dysfunction, normal RV systolic function, MR NR, PH NR	NR	BB	04 y	Asymptomatic
Pica *et al.*[11] (2014)	M, 22 y	Dyspnoea, severity NR	SR, incomplete RBBB, PRWP	Present	LV apex, subepicardial	NR	Basal VS, mid-wall	Moderate LV systolic dysfunction, RV systolic function NR, MR NR, PH NR	Lamin A/C	BB and ACEI. ICD implantation	05 y	Symptoms improvement with mild-moderate LV systolic dysfunction. No ICD interventions. No clinical events
Orsborne *et al.*[14] (2014)	F, 17 y	Chest discomfort, severity NR	NR	Present	LV apex	NR	Mid-VS, mid-wall	LV systolic function NR, RV systolic function NR, MR NR, PH NR	NR	NR	NR	NR

Continued

Table 1 *Continued*

Authors (year)	Sex and age	Clinical presentation	ECG	Multimodality imaging (TTE, CCT, and CMR)					Genetic mutation	Management	Follow-up	Outcomes
				Morphological features				Functional features				
				ILVAH findings[a]	FR	Associated congenital abnormalities	LGE					
Skidan *et al.*[3] (2019)	M, 32 y	Mild dyspnoea	AF	Present	LV apex, subepicardial	NR	NR	Moderate LV systolic dysfunction, RV systolic dysfunction NR, MR NR, PH	—	BB, ACEI, MRA, digitalis, amiodarone, and rivaroxaban. ICD implantation. AF ablation.	2 y	Symptoms and biventricular systolic function improvement. Atrial volumes and PH unchanged. Recurrence of AF (2 episodes) and occurrence of VT (01 episode) successfully treated by ICD.
Dattani *et al.*[5] (2021)	F, 64 y	Severe dyspnoea, APO	LBBB	Present	LV apex, subepicardial	NR	Absent	Moderate LV systolic dysfunction, normal RV systolic function, MR NR, PH NR	—	BB, ACEI, MRA, AC and amiodarone. CRT-P implantation. AV nodal ablation.	15 y	Symptoms improvement, then she developed decompensated HF and AF. Up-titration of drugs and CRT-P implantation. AV nodal ablation due to poorly controlled rapid symptomatic AF.
Marinelli *et al.*[12] (2021)	F, 52 y	Migraine	Type 1 Brugada pattern	Present	LV apex and mid-lateral wall	NR	LV apex and mid-lateral wall	Normal LV systolic function, normal RV systolic function, MR NR, PH NR	NEXN	ILR implantation	30 mo	No VA detected. Asymptomatic. Persistence of Type 1 Brugada pattern. No changes in TTE.

AC, anticoagulation; ACEI, angiotensin-converting enzyme inhibitor; AF, atrial fibrillation; AFl, atrial flutter; APO, acute pulmonary oedema; AS, aortic stenosis; ASD, atrial septum defect; AV, atrioventricular; BB, beta-blocker; CT, cardiac computed tomography; CICU, cardiac intensive care unit; CMR, cardiac magnetic resonance; CRT-P, cardiac resynchronization therapy pacemaker; DC, direct current; ECG, electrocardiogram; F, female; FR, fatty replacement; HF, heart failure; ICD, implantable cardioverter-defibrillator; ILP, internal loop recorder; ILVAH, isolated left ventricular apical hypoplasia; LBBB, left bundle branch block; LGE, late gadolinium enhancement; LV, left ventricular; M, male; MR, mitral regurgitation; MRA, mineralocorticoid receptor antagonist; MV, mechanical ventilation; MVP, mitral valve prolapse; NR, not reported; NSICD, non-specific intraventricular conduction disturbance; PDA, patent ductus arteriosus; PH, pulmonary hypertension; PM, papillary muscles; PRWP, poor R-wave progression; PS, pulmonary stenosis; RAD, right axis deviation; RAE, right atrial enlargement; RBBB, right bundle branch block; RV, right ventricular; RVH, right ventricular hypertrophy; SR, sinus rhythm; TTE, transthoracic echocardiography; TWA, T-wave abnormalities; VA, ventricular arrhythmia; VF, ventricular fibrillation; VS, ventricular septum.

[a]Isolated left ventricular apical hypoplasia findings include spherical left ventricle with deficient apex, anteroapical papillary muscles' origin, and elongated right ventricle wrapping around left ventricle.

[b]Reported but not specified.

exposed.[12] Transthoracic echocardiogram, besides diagnostic features, usually revealed LV systolic dysfunction, mitral regurgitation, and left atrium dilation of variable grade.[1,3–9] Less frequently, it showed right ventricular dysfunction and PH.[3,4,8] Most of these descriptions were met by our patient (*Table 1*).

Definite diagnosis is based on cardiovascular imaging. Cardiac magnetic resonance and cardiac computed tomography were the first imaging modalities being used[1] while TTE was introduced later[4] owing to the ability of the firsts to detect fatty tissue and exclude differential diagnosis. Unequivocal findings have been reported and include constant features such as a truncated spherical left ventricle with bulging of the ventricular septum towards RV, an abnormal origin of papillary muscles from the flattened LV apex, and an elongated RV wrapping around the deficient LV apex. Furthermore, subepicardial fatty replacement at LV apex has been reported in most cases (*Table 1*).

Causes of myocardial fatty replacement include physiologic and pathologic conditions. Non-pathologic myocardial adipose tissue may normally be detected in healthy elderly, more frequently in right ventricle, with subepicardial distribution, normal or thickened myocardium, and non-dilated ventricles. In contrast, cardiac diseases with myocardial fatty infiltration (aside from ILVAH), mainly comprehending arrhythmogenic right ventricular cardiomyopathy (including left-dominant variant), post-myocardial infarction lipomatous metaplasia, and dilated cardiomyopathy, differ from ILVAH by age of presentation, cardiac location, intramyocardial distribution, myocardial thickness, and ventricular size.[15]

Beholding the young age of presentation, the fatty tissue localization at LV apex with subepicardial distribution, the thinned myocardium, the non-dilated left ventricle, and the other ILVAH findings, differential diagnoses were excluded. In addition, myocardial tissue characterization by CMR allows detection of myocardial fibrosis, which has been described in some cases proposing an alternative acquired pathogenic mechanism, like *in utero* infection,[13] and a potential future role for risk stratification.[14]

Specific treatment is currently not available and management relies on close follow-up as well as treatment of complications, as we are doing for our patient. Guideline-directed medical treatment yielded successful symptoms' control at short term in most cases with the longest surveillance being reported at 15 years (*Table 1*). However, disease progression[5] and death[4] have been reported. Only 3 cases reported the use of invasive treatment strategy including AF ablation,[3] implantable cardioverter-defibrillator,[3,11] and cardiac resynchronization therapy pacemaker.[5] Nonetheless, it is unknown if medical or device therapy improves survival in these patients.

Conclusion

Multimodality non-invasive cardiovascular imaging, including TTE and CMR, is pivotal for accurate diagnosis of ILVAH. Close follow-up and treatment of complications (HF, PH, and AF) remain critical as prognosis is uncertain.

Lead author biography

Dr Ricardo Román Carpio is a Peruvian physician and cardiology fellow in Peru at Cayetano Heredia Hospital. He is also a member of the Peruvian Society of Cardiology. He is interested in the field of Cardiovascular Imaging and is currently finishing his training in clinical cardiology at Cayetano Heredia University.

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 authors confirm that written consent for submission and publication of this case report, including images and associated text, has been obtained from the patient in line with COPE guidelines.

Conflict of interest: None declared

Funding: None declared.

References

1. Fernandez-Valls M, Srichai MB, Stillman AE, White RD. Isolated left ventricular apical hypoplasia: a new congenital anomaly described with cardiac tomography. *Heart* 2004;**90**:552–555.
2. Marin C, Sanchez ML, Maroto E, Ossaba S, Ruiz Y, Zabala JI. MR Imaging of isolated left ventricular apical hypoplasia. *Pediatr Radiol* 2007;**37**:703–705.
3. Skidan VI, Kuznetsova T, Pavlyukova EN, Nartsissova GP. Isolated left ventricular apical hypoplasia with myocardial non-compaction: a case report. *Eur Heart J Case Rep* 2019;**4**:1–6.
4. Irving CA, Chaudhari MP. Fatal presentation of congenital isolated left ventricular apical hypoplasia. *Eur J Cardiothorac Surg* 2009;**35**:368–369.
5. Dattani A, Prasad R. Isolated left ventricular apical hypoplasia. *Card Fail Rev* 2021;**7**:e21.
6. Haffajee JA, Finley JJ, Brooks EL, Kuvin JT, Patel AR. Echocardiographic characterization of left ventricular apical hypoplasia accompanied by a patent ductus arteriosus. *Eur J Echocardiogr* 2011;**12**:E17.
7. Moon JI, Jeong YJ, Lee G, Choi JH, Lee JW. Isolated left ventricular apical hypoplasia with infundibular pulmonary and aortic stenosis: a rare combination. *Korean J Radiol* 2013;**14**:874–877.
8. Baroni M, Pedrotti P, Nava S, Giannattasio C, Roghi A. Cardiac magnetic resonance imaging of left ventricular apical hypoplasia in two complex congenital clinical syndromes. *Circ J* 2014;**78**:1507–1509.
9. Vanhecke TE, Decker J, Leonowicz N, Chinnaiyan KM. Isolated left ventricular apical hypoplasia. *Congenit Heart Dis* 2011;**6**:646–649.
10. Limongelli G, Caiazza M. Genetic basis of left ventricular apical hypoplasia. Monaldi Hospital. ClinicalTrials.gov. https://clinicaltrials.gov/ct2/show/study/NCT04339582 (5 March 2022).
11. Pica S, Ghio S, Raineri C, Scelsi L, Turco A, Visconti LO. Mutation of the lamin A/C gene associated with left ventricular apical hypoplasia: a new phenotype for laminopathies? *G Ital Cardiol* 2014;**15**:717–719.
12. Marinelli A, Costa A, Dugo C, Cecchetto A, Lanzoni L, Molon G. A unique case of left ventricle apical hypoplasia presenting with a type 1 Brugada ECG pattern and NEXN mutation. Are they related? *Heart Rhythm Case Rep* 2021;**7**:273–277.
13. Motwani M, Witte KK, Plein S, Greenwood JP. Isolated left ventricular apical hypoplasia evaluated by cardiovascular magnetic resonance and gadolinium enhancement techniques. *J Am Coll Cardiol* 2011;**58**:2355.
14. Orsborne C, Schmitt M. Isolated left ventricular apical hypoplasia, characterized by cardiac magnetic resonance imaging. *Eur Heart J* 2014;**35**:3303.
15. Cannavale G, Francone M, Galea N, Vullo F, Molisso A, Carbone I, *et al.* Fatty images of the heart: spectrum of normal and pathological findings by computed tomography and cardiac magnetic resonance imaging. *Biomed Res Int* 2018;**2018**:5610347.