Isolated left ventricular apical hypoplasia with myocardial non-compaction: a case report

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Background
Isolated left ventricular apical hypoplasia (ILVAH) is a rare congenital cardiac abnormality, which might result in severe symptomatic heart failure (HF) with pulmonary hypertension, atrial fibrillation (AF), or malignant ventricular tachycardia in adults.

Case summary
A 32-years-old man presented with exertional dyspnoea New York Heart Association Class II and persistent AF. Echocardiography and cardiac magnetic resonance showed the presence of (i) spherical remodelling of the left ventricle (LV) with impaired contractile function (three-dimensional ejection fraction, EF 32%); (ii) substitution of apical myocardium by fatty tissue; (iii) abnormal origin of a papillary muscle network; and (iv) an elongated right ventricle, compatible with ILVAH. In addition, non-compacted endomyocardial layer of the LV was observed. Because of a high risk of sudden cardiac death in symptomatic HF patients with reduced EF, an implantable cardioverter-defibrillator was placed which followed by pulmonary vein isolation. After the procedures and restoration of sinus rhythm, the patient demonstrated improvement in HF symptoms and exercise tolerance. This was accompanied by an enhancement of left and right ventricular systolic function by echocardiography. At 6-month, 1, and 2-year follow-up the clinical conditions of the patient and echocardiographic findings remained stable.

Discussion
A rare combination of ILVAH and left ventricular myocardium non-compaction was observed in this young adult who presented with symptomatic HF and persistent AF. The use of consecutive invasive cardiac procedures leads to restoration of sinus rhythm, the improvement of myocardial contractility and clinical manifestation of HF.

Keywords
Isolated left ventricular apical hypoplasia • Myocardial non-compaction • Atrial fibrillation • Implantable cardioverter-defibrillator • Case report

Learning points
• Transthoracic echocardiography and cardiac magnetic resonance are important imaging modalities to diagnose, isolated left ventricular apical hypoplasia (ILVAH) which is a rare congenital cardiac abnormality.
• We reported a case of a combination of ILVAH and left ventricular myocardium non-compaction which manifested as symptomatic biventricular heart failure (HF) and persistent atrial fibrillation.
• The use of consecutive cardiac procedures leads to the restoration of sinus rhythm, prevention of arrhythmias, the enhancement of myocardial contractility and, therefore, improvement of clinical manifestation of HF.

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Introduction

Isolated left ventricular apical hypoplasia (ILVAH) is a rare congenital cardiac abnormality, which was first described by Fernandez-Valls et al.1. To our knowledge, 42 clinical cases of ILVAH were described in the literature.2 The clinical severity may vary from asymptomatic in children to severe symptomatic heart failure (HF) with pulmonary oedema, atrial fibrillation (AF), or malignant ventricular tachycardia (VT) in adults.3–5 The early diagnosis of ILVAH by transthoracic echocardiography (TTE) and cardiac magnetic resonance (CMR) is particularly important due to the difference in the prognosis and treatment strategies of this condition as compared to other types of cardiac disorders with similar clinical manifestations.6–8 Here, we present a case of an adult with ILVAH and LV non-compaction complicated by persistent AF and biventricular HF.

Timeline

| Timeline                  | Details                                                                 |
|---------------------------|-------------------------------------------------------------------------|
| Initial hospitalization   | (i) The patient presented with exertional dyspnoea New York Heart Association Class II and persistent atrial fibrillation (AF). (ii) Transthoracic echocardiography (TTE) demonstrated spherical dilatation of left ventricle (LV) with a thickened, truncated, and akinetic LV apex. The right ventricle appeared elongated and was wrapped around the distal part of the LV. The LV and right ventricle (RV) contractility and systolic function were moderately reduced. Doppler recordings showed signs of elevated LV filling pressure and moderate pulmonary hypertension (PHT). (iii) The cardiac magnetic resonance helped further with visualization of the apical part of the heart and confirmed the diagnosis of left ventricular apical hypoplasia (ILVAH) and LV non-compaction. |
| Consecutive invasive      | (i) A dual-chamber implantable cardioverter-defibrillator (ICD) with passive fixation of the electrode in the apical part of the RV and the atrial electrode in the right atrial appendage with appropriate biventricular pacemakers sensing. (ii) Two months after ICD implantation the pulmonary vein isolation procedure was performed using the three-dimensional-mapping system (CARTO-3). |
| procedures               |                                                                         |
| 6-month follow-up        | (i) Positive clinical evolution with good exercise capacity and stable sinus rhythm under cordarone 200 mg once daily, eplerenone 25 mg/day, rivaroxaban 20 mg/day, and perindopril 4 mg/day. (ii) Transthoracic echocardiography demonstrated substantial improvement in contractile and systolic function of both ventricles. Left and right atrial volumes unchanged and moderate tricuspid regurgitation and PHT were also observed. |
| 1-year follow-up         | The clinical condition of the patient and TTE findings remained stable and similar to the 6-month observations. |
| 15-month follow-up       | (i) The patient presented with complaints of palpitations and chest discomfort. (ii) A paroxysm of AF was documented with a subsequent restoration of sinus rhythm. (iii) One day later a paroxysm of ventricular tachycardia (VT) was detected, sinus rhythm was successfully restored by Antiarrhythmia pacing (ATP). |
| 22-month follow-up       | (i) The patient presented with complaint of chest discomfort. (ii) A paroxysm of the AF with heart rate falling into VT zone and a single unsuccessful attempt of rhythm restoration were recorded. (iii) Sinus rhythm was spontaneously restored the next day. |
| 2-year follow-up         | The clinical condition of the patient remained stable and the echocardiographic parameters did not change substantially as compared to a 1-year follow-up. |

Case presentation

A 32-year-old male with exertional dyspnoea New York Heart Association (NYHA) Class II and persistent AF was admitted to the local hospital for the pulmonary vein isolation (PVI). Baseline medication therapy was with digoxin (0.125 mg/day), bisoprolol (5.0 mg/day), and rivaroxaban (20 mg/day). The baseline therapy was administered during 6 months before hospitalization but without substantial improvements in rate control or HF symptoms. Because of persistent tachycardia, which significantly aggravated the severity of HF symptoms, it was decided to conduct an AF ablation. Blood pressure was 110/65 mmHg; heart rate was irregular at 70–125 b.p.m., with the signs of neck vein distention and peripheral oedema (lower leg oedema). Heart sounds were muffled and the systolic murmur was heard in the second intercostal space at the left sternal border and the apex. Electrocardiogram showed sustained AF which was confirmed by a subsequent Holter monitoring. Transthoracic echocardiography demonstrated spherical deformation (distal) of left ventricle (LV) with a thickened, truncated, and akinetic LV apex. Left ventricular contractility and systolic function were moderately reduced (ejection fraction, EF 32%). The right ventricle (RV) appeared elongated and was wrapped around the distal part of the LV. Right ventricular systolic function was diminished (fractional area change 29%). Doppler recordings showed signs of elevated LV filling pressure (E/e’=18) and moderate pulmonary hypertension (PHT). Table 1 lists the main echo-
RV wrapping around the deficient apex. In addition, non-compacted endomyocardial layer was described in the mid-cavity of anterior and lateral walls of the LV.

After admission to the hospital, the patient received additionally ACE inhibitor (perindopril 4 mg/day) and diuretic (eplerenone 25 mg/day). Based on the clinical and imaging data and because of a high risk of sudden cardiac death (SCD) in symptomatic HF patients with reduced EF, an implantable cardioverter-defibrillator (ICD) was placed which followed by the PVI procedure. First, we implanted a dual-chamber ICD with fixation of the endocardial ventricular defibrillation electrode in the apical part of the RV and the atrial electrode in the right atrial appendage with appropriate biventricular pacemakers sensing. Already after ICD implantation, the patient demonstrated improvement in HF symptoms and exercise tolerance. This was accompanied by an enhancement of left and right ventricular systolic function as assessed by echocardiography (Table 1). Of note, left atrial and right atrial volumes unchanged over the follow-up period and moderate tricuspid regurgitation and PHT were observed (Table 1).

At 6-month follow-up, the clinical condition of the patient and TTE findings remained stable and similar to the 6-month observations. However, after the 15 months of follow-up, the patient presented with complaints of palpitations and chest discomfort. Using the Care Link system, a paroxysm of AF was documented with a subsequent restoration of sinus rhythm through the application of a 23 J electrical shock (Supplementary material online, Figure S1A). One day later a paroxysm of VT (280 b.p.m.) was detected, sinus rhythm was successfully restored by antitachycardia pacing (Supplementary material online, Figure S1b).

After a 22 months follow-up, the patient presented again with a chest discomfort. The Care Link system recorded a paroxysm of AF with heart rate falling into VT zone and a single unsuccessful attempt of rhythm restoration (Supplementary material online, Figure S1c). Sinus rhythm was restored spontaneously the next day.

After a 2-year follow-up, the clinical condition of the patient remained stable and the echocardiographic parameters did not change substantially as compared to a 1-year follow-up (Table 1).

### Table 1 Echocardiographic characteristics of the patient at baseline and follow-up

| Characteristics | Baseline | After ICD | After 6 months | After 12 months | After 24 months |
|-----------------|----------|-----------|----------------|----------------|----------------|
| **2D LV**       |          |           |                |                |                |
| EDV (mL)        | 162      | 152       | 159            | 158            | 155            |
| ESV (mL)        | 107      | 77        | 72             | 77             | 78             |
| Stroke volume (mL) | 55   | 75        | 87             | 81             | 77             |
| Ejection fraction (%) | 34 | 49        | 54             | 51             | 49             |
| Cardiac output (L/min) | 2.8 | 4.9       | 6.4            | 5.2            | 5.0            |
| **3D LV**       |          |           |                |                |                |
| EDV (mL)        | 171      | 155       | 163            | 169            | 165            |
| ESV (mL)        | 116      | 79        | 80             | 77             | 79             |
| Stroke volume (mL) | 55   | 76        | 83             | 92             | 86             |
| Ejection fraction (%) | 32 | 49        | 51             | 54             | 52             |
| **3D RA volumea (mL)** | 85 | 80        | 88             | 100            | 98             |
| **2D RV**       |          |           |                |                |                |
| TAPSE (mm)      | 11       | 14        | 18             | 18             | 21             |
| FAC (%)         | 29       | 41        | 47             | 49             | 51             |
| **3D RA volumea (mL)** | 56 | 46        | 50             | 45             | 44             |
| Doppler data    |          |           |                |                |                |
| E/e' ratio      | 18       | 16        | 12             | 12             | 13             |
| TR degree       | 2        | 2         | 2              | 2              | 2              |
| PG (mmHg)       | 63       | 60        | 60             | 50             | 50             |
| PASP (cm)       | 73       | 70        | 70             | 60             | 60             |

2D, two-dimensional; 3D, three-dimensional; EDV, end-diastolic volume; ESV, end-systolic volume; FAC, fractional area changes; LA, left atrium; LV, left ventricle; PASP, pulmonary artery systolic pressure; PG, pressure gradient; RA, right atrium; RV, right ventricle; TAPSE, tricuspid annular plane systolic excursion; TR, tricuspid regurgitation.

aTest Statistic c=0.6257.
BL ± after ICD: P-value = 0.5758.
BL < after 6 m: P-value = 0.7121.
BL < after 12 m: P-value = 0.1879.
BL < after 24 m: P-value = 0.2871.
Discussion

Because the patients with ILVAH present with non-specific symptoms such as exertion dyspnoea or chest discomfort, the imaging techniques such as TTE and CMR are crucially important for diagnosis of this rare cardiac disorder. ILVAH characterized by (i) spherical remodelling of the LV with impaired contractile function; (ii) substitution of apical myocardium by fatty tissue; (iii) abnormal origin of the papillary muscles from the apex of left ventricle; (iv) three-dimensional: transthoracic echocardiography three-dimensional speckle tracking before surgery; (D) transoesophageal echocardiogram five-chamber view: a truncated left ventricle with spherical remodelling; (E) transoesophageal echocardiogram three-dimensional: aortic valve displacement in non-orthogonal plane; and (F) transthoracic echocardiography three-dimensional: the shape of left ventricle with the deficient left ventricular apex.

Figure 1  Transthoracic echocardiography, transoesophageal echocardiogram two-dimensional-, and three-dimensional echocardiographic images of the patient at baseline: (A) transthoracic echocardiography four-chamber view: bulging of septum towards the right ventricle, elongated right ventricle wrapped around the left ventricular apex; (B) transthoracic echocardiography three-chamber modified view: abnormal origin of the papillary muscles from the apex of left ventricle; (C) three-dimensional: transthoracic echocardiography three-dimensional speckle tracking before surgery; (D) transoesophageal echocardiogram five-chamber view: a truncated left ventricle with spherical remodelling; (E) transoesophageal echocardiogram three-dimensional: aortic valve displacement in non-orthogonal plane; and (F) transthoracic echocardiography three-dimensional: the shape of left ventricle with the deficient left ventricular apex.

Figure 2  Cardiac magnetic resonance images of the patient showing spherical left ventricle with deficient apex formed by the right ventricle, apical fatty infiltration and left ventricle non-compaction.
described a successful implementation of medical and invasive treatment in the patient with a rare combination of ILVAH and LV non-compaction accompanied by a persistent AF and biventricular symptomatic HF. First, we implanted a dual-chamber ICD as this procedure is indicated in patients with significantly reduced LV function (EF <35%) and NYHA Class II or III even if there are no history of VT.14 Moreover, ICD is also recommended in patients with myocardial LV non-compaction due to a high risk of SCD.15 Secondly, after the sequential pulmonary vein isolation (ablation) (PVI/A) procedure, a full recovery of sinus rhythm in our patient leads to further improvement of clinical manifestations and LV function. On the other hand, echocardiographic data at 12-month demonstrated the presence of restrictive type of the LV diastolic function, dilatation, and volume overload of the left heart chambers and PHT, which requires further monitoring and continuation of medical treatment.

In conclusion, TTE and CMR are important imaging modalities to diagnose ILVAH, which determines the clinical diagnosis and treatment strategy. A rare combination of ILVAH and LV myocardium non-compaction was observed in this young adult who presented with symptomatic biventricular HF and persistent AF. The first time in the medical practice, the use of active invasive strategy leads to the restoration of sinus rhythm, prevention of arrhythmias, the enhancement of myocardial contractility and, therefore, improvement of clinical manifestation of HF.

Supplementary material

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

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

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

Conflict of interest: none declared.

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