ESC European Society of Cardiology

European Heart Journal - Case Reports (2020) **4**, 1–6
doi:10.1093/ehjcr/ytz215

CASE REPORT
Congenital heart disease

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

Viktoria I. Skidan [1]*, Tatiana Kuznetsova[2], Elena N. Pavlyukova [3], and Galina P. Nartsissova [4]

[1]Federal Center of Cardiovascular Surgery, 2v Krasnodarskaya str., Khabarovsk 680000, Russian Federation; [2]Research Unit Hypertension and Cardiovascular Epidemiology, KU Leuven Department of Cardiovascular Sciences, University of Leuven, Leuven, Belgium; [3]Cardiology Research Institute, TNRMC, Tomsk, Russian Federation; and [4]National Medical Research Center named after E.N. Meshalkin, Novosibirsk, Russian Federation

Received 2 March 2019; first decision 13 May 2019; accepted 12 November 2019; online publish-ahead-of-print 31 December 2019

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.

* Corresponding author. Tel: +7 (914) 407 62 25, Fax: +7 (4212) 75 88 00, Email: skivi5@yandex.ru

Handling Editor: Matteo Cameli

Peer-reviewers: Domenico D'Amario and Hatem Soliman Aboumarie

Compliance Editor: Mohammed Majid Akhtar

Supplementary Material Editor: Peregrine Green

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.[6–9] Here, we present a case of an adult with ILVAH and LV non-compaction complicated by persistent AF and biventricular HF.

Timeline

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 procedures	(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).
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 Antitachycardia 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 (dilatation) 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 echocardiographic two-dimensional, three-dimensional, and Doppler parameters at the baseline and during the follow-up examinations. Figure *1* shows the TTE images recorded at baseline.

The CMR helped further with visualization of the apical part of the heart (*Figure 2*) and demonstrated the features that characterized ILVAH such as: (i) shortening and spherical remodelling of the LV with impaired contractile function; (ii) invagination of fatty tissue (up to 5 mm) into the myocardium of the defective LV apex; (iii) the anteroapical LV origin of a papillary muscle network; and (iv) an elongated

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 LA volume[a] (mL)	85	80	88	100	98
2D RV					
TAPSE (mm)	11	14	18	18	21
FAC (%)	29	41	47	49	51
3D RA volume[a] (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.
[a]Test Statistic t=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.

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*). Subsequently, two months after ICD implantation the PVI procedure was performed using the three-dimensional-mapping system (CARTO-3). Sinus rhythm is restored by electric pulse therapy 270 J.

After a 6-month follow-up, the clinical condition of the patient was further improved with a good exercise capacity (NYHA Class I) and stable sinus rhythm under cordarone 200 mg/day, eplerenone 25 mg/day, rivaroxaban 20 mg/day, and perindopril 4 mg/day. Although TTE at 6-month still demonstrated the spherical remodelling of the LV, we observed further improvement in contractile and systolic function of both ventricles (*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*).

After a 1-year 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*).

At 22 months, the patient presented again with a chest discomfort. The Care Link system recorded a paroxysm of the 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*).

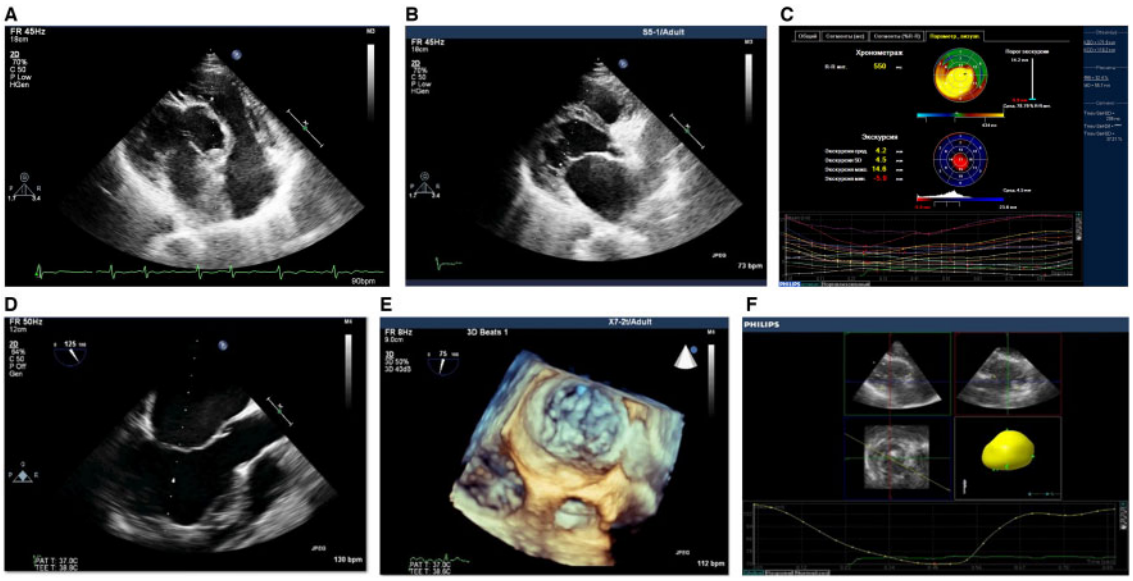

Figure 1 Transthoracic echocardiography, transoesophageal echocardiogram two-dimensional-, and three-dimensional echocardiographic images of the patient at baseline: (*A*) transthoracic echocardiography four-chamber view: bulging of septum towards the right ventricle, elongated right ventricle wrapped 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.

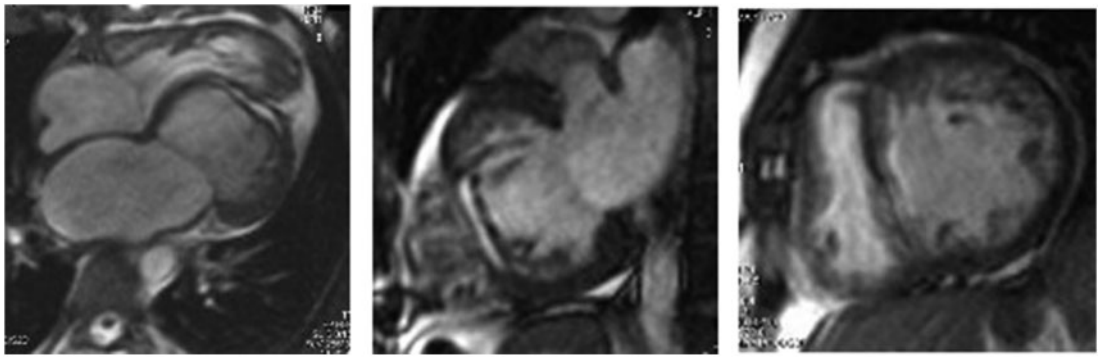

Figure 2 Cardiac magnetic resonance images of the patient showing spherical left ventricle with deficient apex formed by the right ventricle, apical fatty infiltration and left ventricle non-compaction.

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 a papillary muscle network; and (iv) an elongated RV wrapping around the deficient apex. The described cardiomyopathy occurs often in an isolated form, but in some patients, it might be in combination with other congenital malformations and abnormalities.[10–12] Left ventricular apical hypoplasia may also manifest by paroxysmal supraventricular rhythm disturbances, persistent AF, and VT. The combination of ILVAH with other structural heart anomalies determines the severity of the disease.[13] Overall, ILVAH requires dynamic observation even of asymptomatic patients and execution of surgical treatment tactics in addition to drug therapy.

We consider this case as a sporadic one since all first-degree relatives were without any signs of congenital heart defects. There was also no familial history of any cardiac pathology. Hereby, we

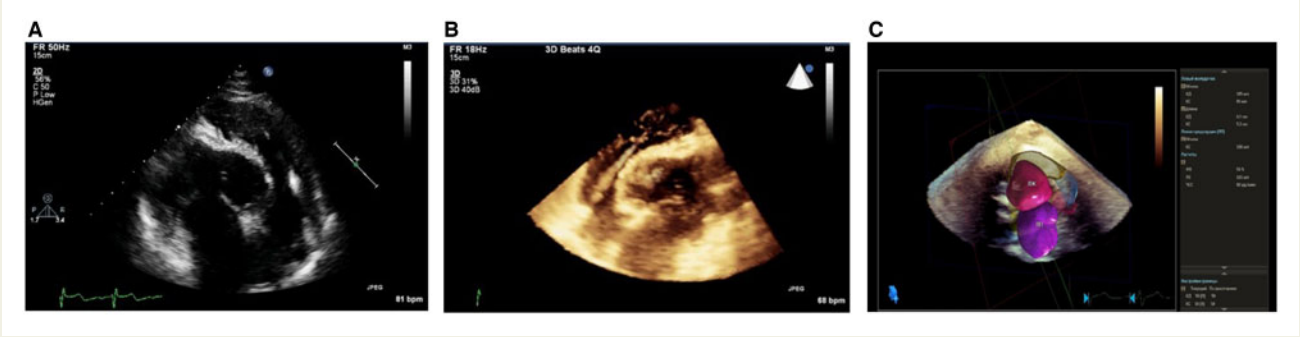

Figure 3 Transthoracic echocardiography two-dimensional and three-dimensional echocardiographic images of the patient after surgical treatment at 12 months: (*A*) transthoracic echocardiography four-chamber view: sinus rhythm, bulging of septum towards the right ventricle, implantable cardioverter-defibrillator-electrode in the right ventricle; (*B*) transthoracic echocardiography three-dimensional heart model: the shape of normally functioning left ventricle and dilated left atrium; and (C) transthoracic echocardiography three-dimensional speckle tracking after surgery.

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.

Lead author biography

Skidan I. Viktoria (Cand Sci (Med), PhD) was a fellow cardiologist and an echocardiographer at the Far Eastern State Medical University, Russian Federation, in 2001–04. In 2005–09, she was an Associate Professor at the Far Eastern State Medical University and was appointed as a researcher and a cardiologist at Echo Research lab, Cardiology specialization training, Salzburg, Austria. In 2009–10, she was a visiting researcher at Cleveland Clinic Foundation, OH, USA, Department of Cardiovascular Imaging, educational grant of the Faculty Development of Fulbright Program. In 2011–19, she is a fellow cardiologist and an echocardiographer at the Federal Center of Cardiovascular Surgery, Khabarovsk, Russian Federation. Her research fields include 3D TTE, TEE, and myocardial 2D/3D deformation imaging connected to HFrEF, right ventricular function and cardiomyopathy. Her scientific outputs include 97 papers and abstracts in peer-reviewed journals, two books as a co-editor, and abstracts at international congresses (EuroEcho, ESC, and ASE). She holds memberships of ESC, EACVI, HF, ASE, and RSC.

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.

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. Skidan VI, Pavlyukova EN, Narcissova GP. Isolated left ventricular apical hypoplasia, rare inborn cardiomyopathy. *Sib Med ž (Tomsk)* 2018;**33**:21–26 (in Russian).
3. Sani ZA, Vojdanparast M, Rezaeian N, Seifi A, Tehrani SO, Nezafati P. Left ventricular apical hypoplasia: Case report on cardiomyopathy and a history of sudden cardiac death. *ARYA Atheroscler* 2016;**12**:50–54.
4. Mirdamadi A, Ashrafi S. Isolated left ventricular apical hypoplasia: manifestations and different echocardiography features. *Iran Red Crescent Med J* 2016;**18**: e26065.
5. 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.
6. Orsborne C, Schmitt M. Isolated left ventricular apical hypoplasia, characterized by cardiac magnetic resonance imaging. *Eur Heart J* 2014;**35**: 3303.

7. Flett AS, Elliott PM, Moon JC. Images in cardiovascular medicine. Cardiovascular magnetic resonance of isolated left ventricular apical hypoplasia. *Circulation* 2008; **117**:504–505.
8. Patrianakos AP, Protonotarios N, Zacharaki A, Tsatsopoulou A, Parthenakis FI, Vardas PS. Isolated left ventricular apical hypoplasia: a newly recognized unclassified cardiomyopathy. *J Am Soc Echocardiogr* 2010;**3**:1336.
9. Hong SA, Kim YM, Lee HJ. Three-dimensional endo-cardiovascular volume-rendered cine computed tomography of isolated left ventricular apical hypoplasia: a case report and literature review. *Korean J Radiol* 2016;**17**:79–82.
10. Zhao Y, Zhang J, Zhang J. Isolated left ventricular apical hypoplasia with right ventricular outflow tract obstruction: a rare combination. *Ann Noninvasive Electrocardiol* 2015;**20**:502–505.
11. 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.
12. Chaowu Y, Xin S, Shihua Z, Jianrong L, Hao W. Complete transposition of the atrioventricular valves associated with left ventricular apical hypoplasia. *Circulation* 2011;**124**:538–539.
13. Wang T, Chen H, Ma X, Fan Z, Fan Z. Isolated left ventricular apical hypoplasia: a case report and literature review. *J Gen Pract* 2016;**4**:4.
14. Epstein AE, DiMarco JP, Ellenbogen KA, Estes NA 3rd, Freedman RA, Gettes LS. 2012 ACCF/AHA/HRS focused update incorporated into the ACCF/AHA/HRS 2008 guidelines for device-based therapy of cardiac rhythm abnormalities: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines and the Heart Rhythm Society. *Circulation* 2013;**127**:e283–e352.
15. Priori SG, Blomström-Lundqvist C, Mazzanti A, Blom N, Borggrefe M, Camm J, Elliott PM, Fitzsimons D, Hatala R, Hindricks G, Kirchhof P, Kjeldsen K, Kuck KH, Hernandez-Madrid A, Nikolaou N, Norekvål TM, Spaulding C, Van Veldhuisen DJ; ESC Scientific Document Group. 2015 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death: The Task Force for the Management of Patients with Ventricular Arrhythmias and the Prevention of Sudden Cardiac Death of the European Society of Cardiology (ESC). Endorsed by: Association for European Pediatric and Congenital Cardiology (AEPC). *Eur Heart J* 2015;**36**:2793–2867.