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Case Report

A late-onset arrhythmogenic right ventricular cardiomyopathy unveiled by supraventricular tachycardia: Case report ☆

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ABSTRACT

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a rare myocardial genetic disease that leads to heart failure and sudden cardiac death. The diagnosis of ARVC remains challenging given its variable presentation. A 66-year-old woman was admitted to our cardiac intensive care unit for the management of a left heart failure flare-up with an atrial flutter, which remains an atypical mode of the revelation of arrhythmogenic right ventricular cardiomyopathy (ARVC) retained based the association of genetic predisposition, the presence of repolarization abnormalities on electrocardiogram (EKG), the occurrence of an episode of sustained ventricular tachycardia and imaging data. ARVC is a condition with varying expressions, severity, and progression. Early detection, careful monitoring, and appropriate management are crucial in improving outcomes for individuals with ARVC.

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Introduction

Arrhythmogenic right ventricular cardiomyopathy (ARVC), also known as arrhythmogenic right ventricular dysplasia (ARVD), is a rare genetic myocardial disease with predominantly autosomal dominant transmission and primarily in the right ventricle (RV). It was first described in the 1970s [1]. ARVC was incorporated into the European Cardiomyopathy Group classification in 1994 [2]. It is characterized histologically by the progressive replacement of normal myocardium by fibro-fatty tissue, leading to global or regional ventricular dysfunction, and predisposing to life-threatening ventricular arrhythmias irrespective of the severity of systolic ventricular dysfunction [3]. The diagnosis of ARVD is challenging and is based on a scoring system using minor and major criteria: the Modified Task Force Criteria (TFC) [2]. Supraventricular tachycardia (SVT) is common in ARVC, with atrial fibrillation and atrial flutter being the most prevalent [4]. We report the case of ARVC with a fast atrial flutter, revealing a late onset ARVC subsequently confirmed by cardiac MRI.

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Case presentation

We report the case of a 66-year-old female patient with only obesity as a cardiovascular risk factor, without any notable personal chronic disease. Her son has been followed for an ARVC with an identified Plakophilin 2 mutation. There was no history of alcohol consumption or illicit drug use. The patient was admitted to our cardiac intensive care unit for the management of paroxysmal palpitations dating back 3 months, which became permanent associated with dyspnea stage IV of the New York Heart Association (NYHA) classification. She reported no previous angina, syncope, or neurological signs. On initial physical examination, the patient was apyretic (36.5°C), tachycardic at 150 beats per minute, with blood pressure at 113/73 mmHg, a respiratory rate of 25 cycles per minute, and a saturation of oxygen in ambient air at 88%. Pulmonary auscultation revealed bilateral basithoracic crackling rales. The EKG on admission presented an atrial flutter with 2:1 atrioventricular conduction and a mean ventricular rate of 150 beats per minute (Fig. 1).

The biological analysis revealed an increased plasma D-dimer level (2990 ng/mL), hepatic cytolysis with an Aspartate Aminotransferase (ASAT) level of 435 mUI/l, and an Alanine Aminotransferase (ALAT) level of 405 mUI/l, an elevated C-reactive protein (CRP) level of 93.7 mg/L, a creatinine level of 12.66 mg/L (Glomerular filtration rate of 44,97 ml/min/1,73m^2), a blood potassium level of 3.8 mmol/l, a hemoglobin level of 11.9g/dL, and platelets of 179000/mm^3.

Transthoracic echocardiography (TTE) showed multiple findings. The RV was dilated (ratio RV/LV = 1) with akinesia of its lateral wall that was hyper-trabeculated with thickened trabeculations. The thickest one measured 5 mm. There was an RV systolic dysfunction (RV shortening fraction at 8 %, tricuspid annular plane systolic excursion à 12 mm) and a flat septum. The left ventricle (LV) wasn't dilated or hypertrophied, with preserved global and segmental systolic function and an ejection fraction (EF) of 63%. The LV filling pressures were elevated. Additionally, the pulmonary artery trunk was dilated measuring 31 mm and there was minimal pulmonary insufficiency with systolic pulmonary arterial pressure at 70 mmHg. The pulmonary resistances calculated were 2.9 WU. Both left and right atrium were dilated, and the inferior vena cava was noncompliant and measured 22 mm. The pericardium appeared normal without any signs of fluid accumulation. A contrast-enhanced thoracic scan was performed, which showed a bilateral pleural effusion without any signs in favor of pulmonary embolism.

To restore the patient's sinus rhythm, electrical cardioversion was performed following transesophageal echocardiography, which confirmed the absence of any thrombus. After a single electric shock, the patient restored sinus rhythm. The postelectroshock EKG showed a regular sinus rhythm with a heart rate of 50 beats per minute. Inverted T-waves were observed in all leads except AvL and DI, with a prolonged QT interval (QTc 480 ms) and no epsilon wave (Fig. 2). Approximately 2 hours after the cardioversion, the patient experienced a recurrence of supraventricular tachycardia with a heart rate of 165 beats per minute. The initial evolution under antiarrhythmic drugs was marked by an alternation between a rapid atrial flutter rhythm and a slower regular sinus rhythm, accompanied by multiple ventricular extrasystoles. During the patient's stay in our cardiac intensive care unit, a brief and well-tolerated episode of sustained ventricular tachycardia occurred, with a heart rate of 240 beats per minute. This episode was followed by a spontaneously reduced torsade de pointes (Fig. 3).

The diagnosis of arrhythmogenic right ventricular cardiomyopathy was established based on the presence of 4 ma-

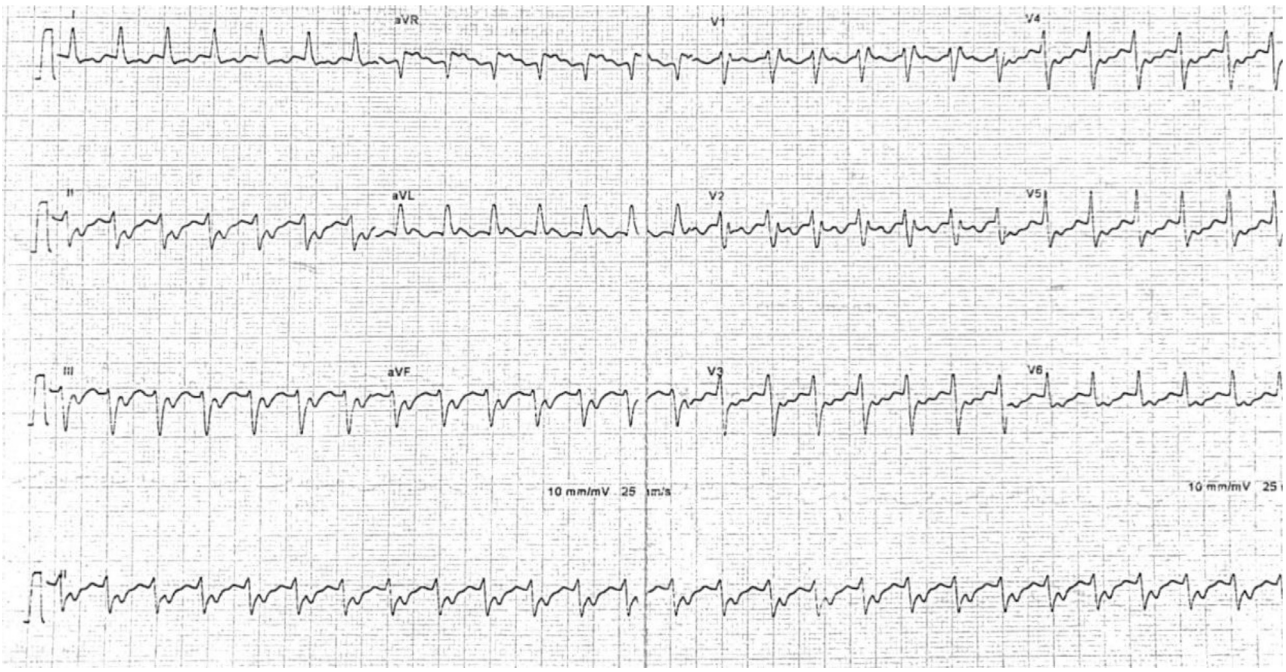

Fig. 1 – EKG at admission showing atrial flutter with nonvariable AV conduction with a heart rate of 150 bpm.

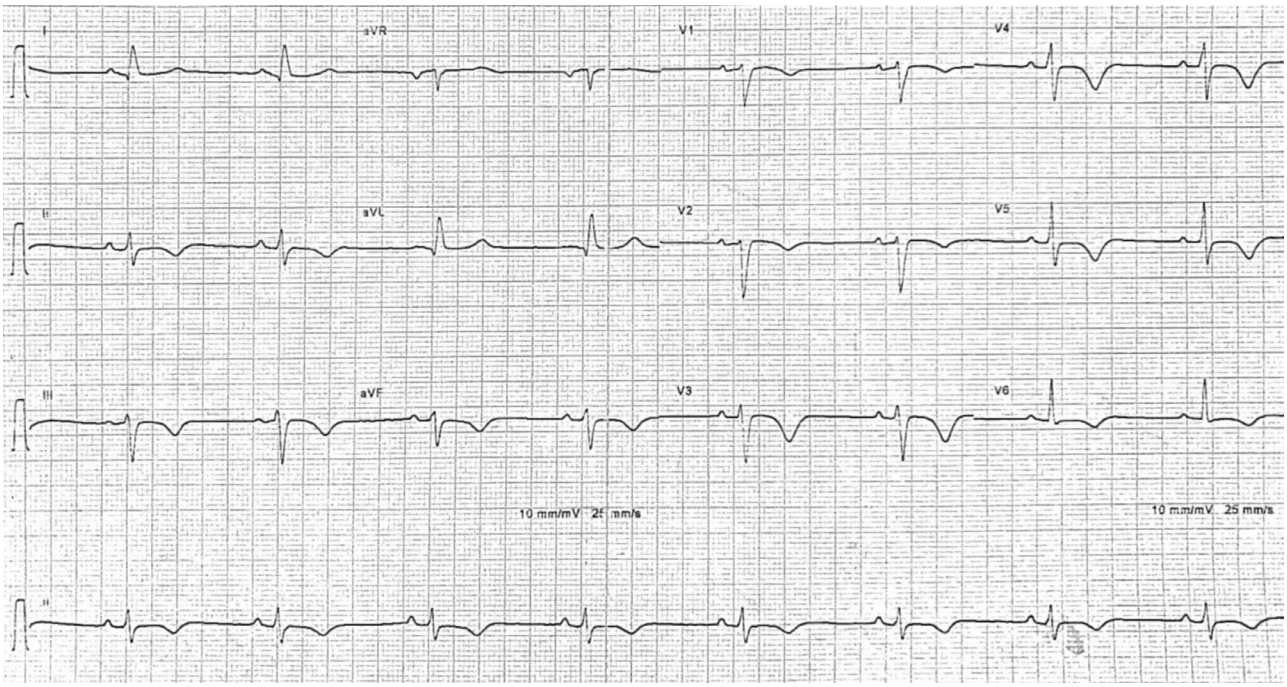

Fig. 2 – EKG after restoring a normal sinus rhythm with a heart rate of 50 bpm by synchronized cardioversion.

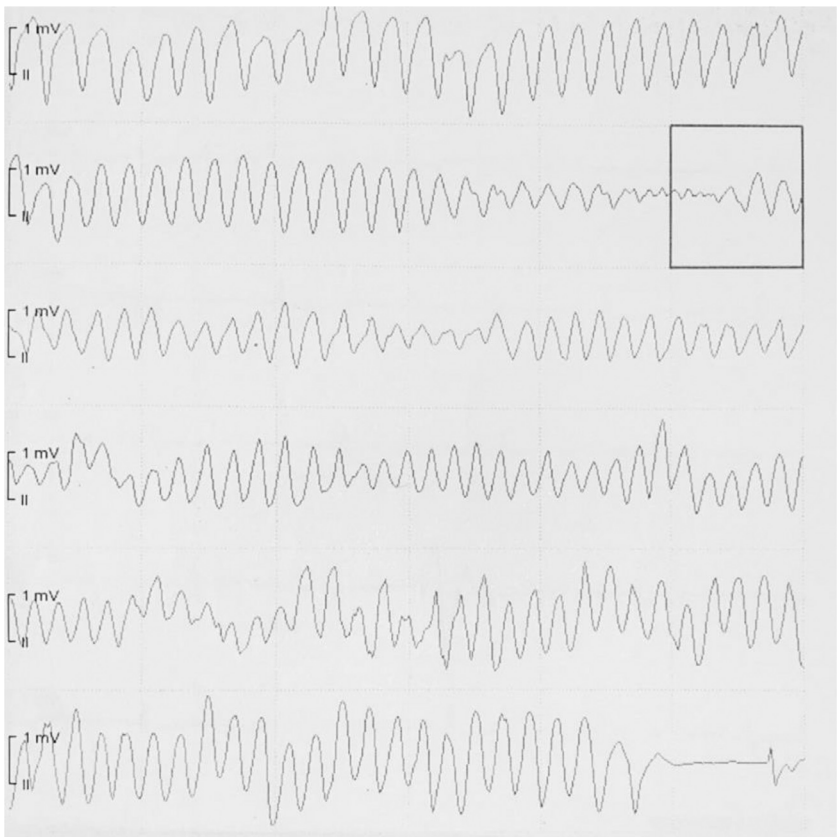

Fig. 3 – Telemetric monitoring in the cardiac intensive care unit: Torsade de pointes.

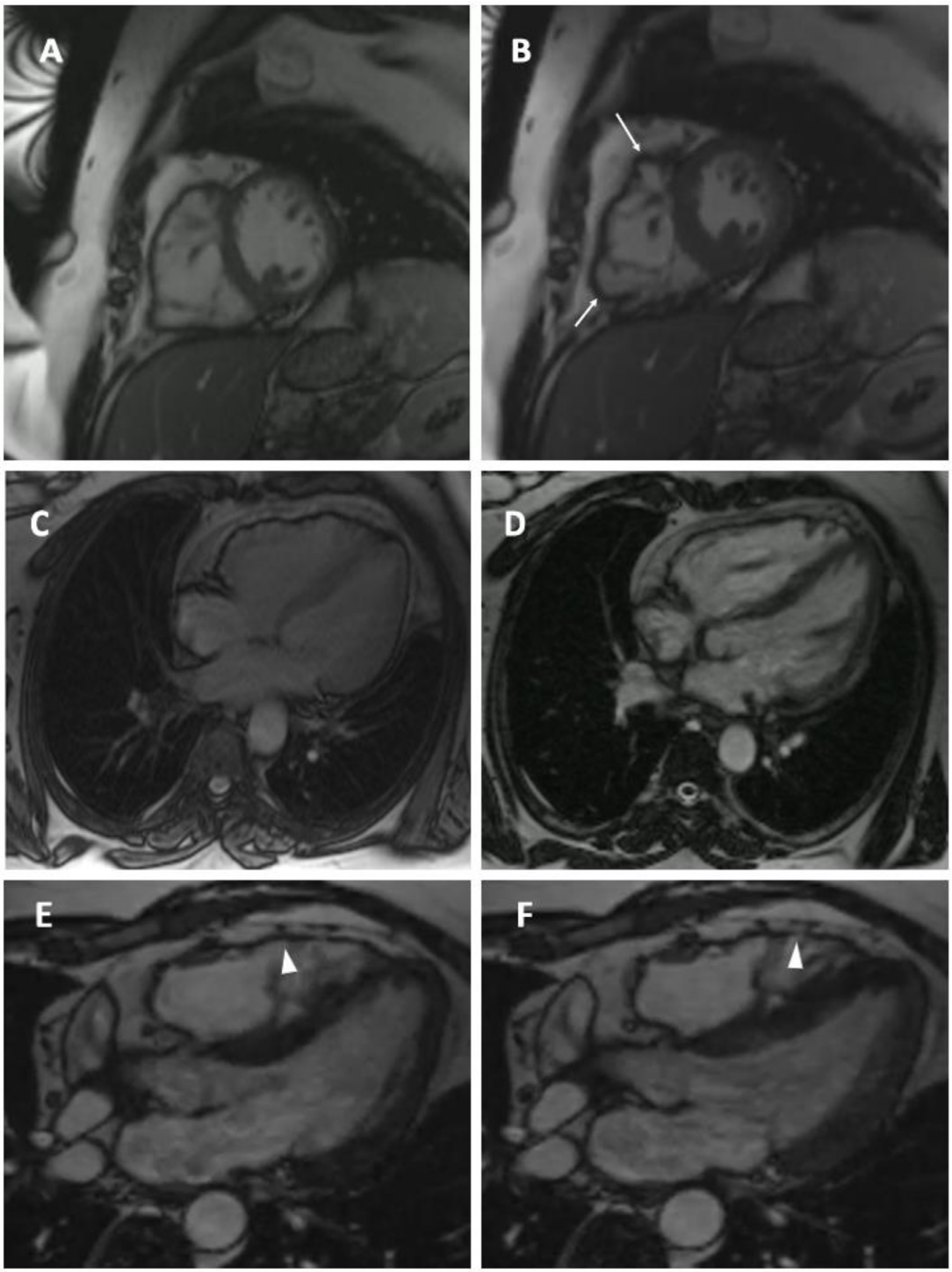

Fig. 4 – Cardiac MRI of our patient: (A) diastole; (B) systole: Short axis views showing a dilated RV with multiple aneurysms of the RV wall (Arrows). (C) 4 chamber view; (D-F) 5 chamber views showing RV dilatation and prominent RV trabeculations.

jor criteria in the modified TASK Force Criteria. These criteria included a genetic predisposition (the patient's son had an identified genetic mutation of ARVC), repolarization abnormalities observed on the electrocardiogram (inversed T-waves without bundle branch block), episodes of sustained ventricular tachycardia, and echocardiographic findings such as akinesia of the lateral wall of the right ventricle and increased size of the right ventricular outflow tract measured at 37 mm (greater than the threshold of 36 mm) along with a reduced right ventricular fraction at 8%.

Later, the patient underwent a cardiac MRI to search for further radiological diagnostic criteria to reinforce the retained diagnosis. It revealed other major radiological criteria, namely a regional RV akinesia, an RV end-diastolic volume of 127

mL/m^2 ($\geqslant$100 mL/m^2), and an altered RV ejection fraction of 25% ($\leq$ 40%). MRI also revealed multiple aneurysms, in the infundibular region, in the lateral wall, and the apex of the RV, with a "stack of plates" appearance (Fig. 4). Then, the patient was referred to a specialized center for the placement of an implantable cardioverter-defibrillator (ICD) and was advised that her children must be screened for ARVD.

Discussion

ARVC is one of the most challenging hereditary cardiomyopathies, marked by progressive fibro-fatty infiltration replacing the myocytes, mainly in the RV but also affecting the LV and, according to some evidence, the atrial chambers. This is how recent guidelines came up with the term arrhythmogenic cardiomyopathy (ACM), which is a development of the original term ARVC. The diagnosis of ARVC is difficult due to the absence of a uniform and specific presentation [5–7]. There are many genes associated with ARVC. The most significant genes are related to the desmosome structure, such as plakophilin 2, desmoglein 2, and desmoplakin [8,9]. ARVC occurs in adolescents and young adults between the ages of 20 and 40 but is rarely seen before the age of 10. It's most often diagnosed during the evaluation of ventricular arrhythmias, or a systematic family work-up after the diagnosis of an index case [10]. Yet, the systematic screening wasn't carried out after genetic confirmation of our patient's son's case, and, the initial presentation with supraventricular arrhythmias, as was in our case, is much less common. Certainly, ventricular rhythm disorders are the most frequent and characteristic of ARVC and are generally the cause of sudden death. That said, supraventricular arrhythmias have also been linked to ARVD ever since this cardiomyopathy was first described [3,5,11].

Atrial tachyarrhythmias, such as atrial fibrillation and atrial flutter, occur in 14% of patients with ARVC and may appear in an early stage before ventricular arrhythmias or in the late stage of the disease [12]. In our case, the electrical evolution was fast. The patient presented during her stay in our cardiac intensive care unit with a rapid atrial flutter reduced to sinus rhythm with a progression marked by the onset of ventricular rhythm disorders: ventricular premature beats followed by nonsustained ventricular tachycardia and then a spontaneously reduced torsade de pointes. For the occurrence of atrial arrhythmias at an early stage of the disease, studies suggest the possibility of direct involvement of the right atrium in the pathological process, while for their late appearance with or after the occurrence of ventricular arrhythmias, the underlying pathogenesis of atrial vulnerability is secondary to the progression of the right ventricular disease [6,13]. An observational long-term study suggests that the presence of atrial fibrillation and atrial flutter is associated with an adverse clinical outcome (inappropriate shocks, heart transplantation, cardiac death) in patients with ARVC [5]. Atrial remodeling and atrial arrhythmias can increase the risk of thromboembolic complications and even sudden death [14]. Inverted T waves in leads V1-V2 are one of the main repolarization abnormalities described in ARVC. The appearance of inverted T waves in lateral and inferior leads suggests LV involvement [8]. In our case, after obtaining a sinus rhythm, we noted the presence of inverted T waves in almost all leads, anterior, lateral, and inferior. It's important to mention that conduction abnormalities have also been reported. Adipose tissue in ARVC infiltrates the conduction system tissue [8,15].

The expression, severity, and progression of the disease vary widely among patients, even within the same family [12]. Although a definitive diagnosis is typically yielded through histological study obtained by biopsy, which is rarely performed. Instead, indirect evidence is gathered through major and minor diagnostic criteria. The Modified Task Force Criteria (TFC) revised in 2010, while specific, still lack sensitivity, particularly in the early stages of the disease and among athletes [2,16,17]. False positives are common in athletes because of the similarities between the electrical profile of healthy athletes and the classic characteristics of ARVC [17]. The modified Task Force Criteria for the diagnosis of ARVC are based on the analysis of family history, repolarization abnormalities, arrhythmias, imaging data, and the histological studies of endomyocardial biopsies [18].

Radiological diagnostic criteria established by an international task force in 2010 include echocardiography, MRI, and RV angiography parameters. Echocardiography is used for the initial evaluation due to its wide availability and valuable diagnostic orientation. MRI, which provides the best ability to assess the detailed morpho-functional and tissue characterization of both RV and LV, has become the imaging technique of choice, though it should never be used alone for ARVC diagnosis. Several signs are highly suggestive of ARVC such as regional wall motion abnormalities (RV akinesia, dyskinesia, or bulging), RV dilatation and/or dysfunction, and RV outflow tract enlargement. While fibrofatty infiltration and late gadolinium enhancement (LGE) in both ventricles are well described in ARVC patients, these features remain nonspecific. Cardiac MRI also allows characterization of the disease phenotype, including dominant right, biventricular, and dominant left forms [19–22].

The management of patients with ARVC remains somewhat complex, with 2 key objectives: improving symptoms and preventing complications. Patients with ARVC may have a good prognosis unless ventricular arrhythmia becomes problematic with a risk of sudden death. ICD implantation should be offered to all patients with ARVC, as part of primary or secondary prevention, to reduce the risk of future sudden cardiac [22,23]. The practice of moderate and/or high-intensity exercise, including competitive sports, is not recommended in individuals with ARVC. Physical exercise increases the risk of serious rhythm disorders and sudden death promoted by adrenergic stress and represents one of the major factors in disease evolution [17,22]. For the treatment of atrial arrhythmias in patients with ARVC, catheter ablation is a reliable and secure alternative approach to antiarrhythmic drugs for the maintenance of sinus rhythm, with overall long-term success rates that are comparable to those reported in the general population [24]. Oral anticoagulation to reduce the risk of thromboembolic events and stroke is recommended in patients with ARVD and AF or atrial flutter with a CHA_2DS_2-VASc score $\geq$2 in men or $\geq$3 in women and should be considered with a CHA2DS2-VASc score of 1 in men or of 2 in women [22].

Conclusion

Although uncommon, ARVC is an inherited, life-threatening cardiomyopathy. It is crucial to consider ARVC in both young people and older age groups. Significant effort must be dedicated to collecting various diagnostic criteria—clinical, electrical, and imaging- since no single modality can definitively diagnose ARVC.

Ethical approval

Ethics approval was not obtained because this manuscript is a case report.

Patient consent

Written informed consent was obtained from the patient.

REFERENCES

[1] Marcus FI, Fontaine GH, Guiraudon G, Frank R, Laurenceau JL, Malergue C, et al. Right ventricular dysplasia: a report of 24 adult cases 1982. http://ahajournals.org

[2] Gandjbakhch E, Redheuil A, Pousset F, Charron P, Frank R. Clinical diagnosis, imaging, and genetics of arrhythmogenic right ventricular cardiomyopathy/dysplasia: jacc state-of-the-art review. J Am Coll Cardiol 2018;72(7):784–804. doi:10.1016/j.jacc.2018.05.065.

[3] Nakazato Y, Nakata Y, Tokano T, Ohno Y, Hisaoka T, Sumiyoshi M, et al. A case of arrhythmogenic right ventricular dysplasia with atrial flutter. Jpn Heart J. 1994;35(5):689–94. doi:10.1536/ihj.35.689.

[4] Müssigbrodt A, Knopp H, Efimova E, Weber A, Bertagnolli L, Hilbert S, et al. Supraventricular arrhythmias in patients with arrhythmogenic right ventricular dysplasia/cardiomyopathy associate with long-term outcome after catheter ablation of ventricular tachycardias. Europace 2018;20(7):1182–7. doi:10.1093/europace/eux179.

[5] Saguner AM, Ganahl S, Kraus A, Baldinger SH, Medeiros-Domingo A, Saguner AR, et al. Clinical role of atrial arrhythmias in patients with arrhythmogenic right ventricular dysplasia. Circ J 2014;78(12):2854–61. doi:10.1253/circj.CJ-14-0474.

[6] Klein C, Souissi Z, Brigadeau F, Lacroix D. Arrhythmogenic right ventricular cardiomyopathy and left atrial tachycardia: a case report. Eur Heart J Case Rep 2017;1(2):2–3. doi:10.1093/ehjcr/ytx012.

[7] Arbelo E, Protonotarios A, Gimeno JR, Arbustini E, Barriales-Villa R, Basso C, et al. 2023 ESC Guidelines for the management of cardiomyopathies. Eur Heart J 2023;44(37):3503–626. doi:10.1093/eurheartj/ehad194.

[8] Nunes De Alencar Neto J, Baranchuk A, Bayés-Genís A, Bayés De Luna A. Arrhythmogenic right ventricular dysplasia/cardiomyopathy: an electrocardiogram-based review. Europace 2018;20(FI1):2–3. doi:10.1093/europace/eux202.

[9] Cho Y. Arrhythmogenic right ventricular cardiomyopathy. J Arrhythm 2018;34(4):356–68. doi:10.1002/joa3.12012.

[10] Lui CY, Marcus FI, Sobonya RE. *Letter to the Editor arrhythmogenic right ventricular dysplasia masquerading as peripartum cardiomyopathy with atrial flutter, advanced atrioventricular block and embolic stroke*.; 2002. www.karger.comwww.karger.com/journals/crd.

[11] Ito K, Iwasaki YK, Fujimoto Y, Oka E, Takahashi K, Tsuboi I, et al. Massive right atrial thrombus formation followed by an atrial flutter with 1:1 atrioventricular conduction in a patient with arrhythmogenic right ventricular cardiomyopathy. Intern Med 2016;55(16):2213–17. doi:10.2169/internalmedicine.55.5520.

[12] Ardan M, Saguner M, Corinna Brunckhorst M, Firat Duru M. Atrial arrhythmias in arrhythmogenic cardiomyopathy: at the beginning or at the end of the disease story? copie. Circ J 2014:3 Published online.

[13] Chu AF, Zado E, Marchlinski FE. Atrial arrhythmias in patients with arrhythmogenic right ventricular Cardiomyopathy/dysplasia and ventricular tachycardia. Am J Cardiol 2010;106(5):720–2. doi:10.1016/j.amjcard.2010.04.031.

[14] Wu L, Guo J, Zheng L, Chen G, Ding L, Qiao Y, et al. Atrial remodeling and atrial tachyarrhythmias in arrhythmogenic right ventricular cardiomyopathy. Am J Cardiol 2016;118(5):750–3. doi:10.1016/j.amjcard.2016.06.003.

[15] Caglar N, Pamir G, Km-Al T, Ahmet I, Candan S, Kumbasar A. Right ventricular cardiomyopathy similar to Uhl's anomaly with atria1 flutter and Complete AV Block. Int J Cardiol 1993;38:199–201.

[16] Amin A, Bailey N, Warren A, Reddy B. Non-sustained ventricular tachycardia as a presentation of arrhythmogenic right ventricular cardiomyopathy. Cureus 2023;15(5) Published online. doi:10.7759/cureus.38620.

[17] Gasperetti A, James CA, Cerrone M, Delmar M, Calkins H, Duru F. Arrhythmogenic right ventricular cardiomyopathy and sports activity: From molecular pathways in diseased hearts to new insights into the athletic heart mimicry. Eur Heart J 2021;42(13):1231–43. doi:10.1093/eurheartj/ehaa821.

[18] Sayed A, Pal S, Poplawska M, Aronow Ws, Frishman WH, Fuisz A, et al. Arrhythmogenic right ventricular cardiomyopathy diagnosis. Cardiol Rev 2020;28(6):319–24 Published online. doi:10.1097/CRD.0000000000000292.

[19] Cipriani A, Mattesi G, Bariani R, Cecere A, Martini N, Michieli L, et al. Cardiac magnetic resonance imaging of arrhythmogenic cardiomyopathy: evolving diagnostic perspectives. Eur Radiol. Published online 2021. doi:10.1007/s00330-022-08958-2/Published.

[20] Malik N, Mukherjee M, Wu KC, Zimmerman SL, Zhan J, Calkins H, et al. Multimodality imaging in arrhythmogenic right ventricular cardiomyopathy. Circ Cardiovasc Imaging 2022;15(2):E013725. doi:10.1161/CIRCIMAGING.121.013725.

[21] Murphy DT, Shine SC, Cradock A, Galvin JM, Keelan ET, Murray JG. Cardiac MRI in arrhythmogenic right ventricular cardiomyopathy. Am J Roentgenol 2010;194(4):299–303. doi:10.2214/AJR.09.3450/SUPPL_FILE/FIG_S7.MOV.

[22] Arbelo E, Protonotarios A, Gimeno JR, Arbustini E, Barriales-Villa R, Basso C, et al. 2023 ESC Guidelines for the management of cardiomyopathies: developed by the task force on the management of cardiomyopathies of the European Society of Cardiology (ESC). Eur Heart J 2023;44(37):3503–626. doi:10.1093/eurheartj/ehad194.

[23] Khan Z, Abumedian M, Yousif Y, Gupta A, Myo SL, Mlawa G. Arrhythmogenic right ventricular cardiomyopathy in a patient experiencing out-of-hospital ventricular fibrillation arrest twice: case report and review of the literature. Cureus 2022;14(1):e21457. Published online January 20. doi:10.7759/cureus.21457.

[24] Gasperetti A, James CA, Chen L, Schenker N, Casella M, Kany S, et al. Efficacy of catheter ablation for atrial arrhythmias in patients with arrhythmogenic right ventricular cardiomyopathy—a multicenter study. J Clin Med 2021;10:4962. doi:10.3390/jcm10214962.