CASE STUDY

Navigating the Uncommon: Case Report of Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Successfully Managed with Implantable Cardioverter Defibrillator (ICD)

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ABSTRACT

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a rare genetic disorder known for its role in sudden cardiac death and primarily affects the right ventricle, leading to ventricular arrhythmias (VA). In a documented case, a 35-year-old patient experienced palpitations, and the detection of epsilon waves in the right precordial leads on an electrocardiogram (ECG) raised suspicions of ARVC. Further diagnostic tests, including a 24-hour Holter monitor showing over 500 ventricular extrasystoles and a cardiac MRI indicating right ventricular dyskinesia with transmural late gadolinium enhancement in the apical right ventricle, supported this suspicion. Additionally, an electrophysiologic study captured spontaneous polymorphic ventricular tachycardia and induction ventricle fibrillation, underscoring the need for an implantable cardioverter defibrillator. This case emphasizes the importance of recognizing ECG anomalies, timely further investigation, and appropriate therapeutic measures in managing ARVC, highlighting the vital role of combining cardiac, electrophysiological, and imaging assessments in the risk stratification and management of ARVC.

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Introduction

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an uncommon hereditary cardiomyopathy. The incidence rate of ARVC is estimated to be between 1 in 2500 and 1 in 5000 individuals.[1] Prompt implementation of pharmacotherapy and device therapy may aid in mitigating the risk of arrhythmic events or sudden cardiac death (SCD). It's always interesting to find relatively uncommon diseases with common symptoms, especially when they appear with diagnosis challenges. Here, we provide a case of a young adult who had common complaints of palpitations, which were identified as ARVC and required implanted cardioverter defibrillator (ICD) therapy.

Case

A 35-year-old man with palpitations came into the cardiology clinic. The palpitation has been occurring intermittently for the past twelve months.

There was no medical history, either personal or familial, of unexplained syncope or cardiac arrest. The electrocardiogram (ECG) during his visit revealed epsilon waves in the right precordial leads (**Figure 1**). The results of an echocardiography examination revealed a slightly dilated right ventricle (RV) and right atrium (RA); the remaining cardiac chamber measurements are within normal ranges. An ARVC was suspected, and the patient was recommended to be hospitalized for further investigation and treatment. A holter monitoring was initiated, which revealed a persistent premature ventricular contraction (PVC) rate exceeding 500 over the course of twenty-four hours. Further investigation using a cardiac MRI (CMRI) revealed dyskinesia in the RV and transmural late gadolinium enhancement (LGE) in the apical RV.

An electrophysiology study (EPS) was arranged for the patient. Regardless of the induced ventricle fibrillation (VF), a spontaneous polymorphic ventricular tachycardia (VT) was seen during EPS. These data validated the ARVC diagnosis and indicated the need for implanted cardioverter defibrillators. A dual-chamber implantable cardioverter defibrillator (ICD) was successfully implanted, with no adverse events. Subsequently, the patient was released with a prescription once daily 5mg of bisoprolol.

Discussion

ARVC is a hereditary heart condition characterized by abnormal heart rhythms in the presence of functional and structural abnormalities of the right ventricle. These abnormalities are caused by the partial or complete replacement of heart muscle cells with fatty and fibrous tissue.[2] However, diagnosing ARVC is difficult since it presents with diverse and nonspecific symptoms. This condition is responsible for causing sudden mortality in 30% of young people and 5% of those under the age of 65.[3]

Study found that around 67% of persons diagnosed with ARVC have palpitations, whereas 32% experience syncope, 27% experience unusual chest pain, 6% experience RV failure, and 6% may not

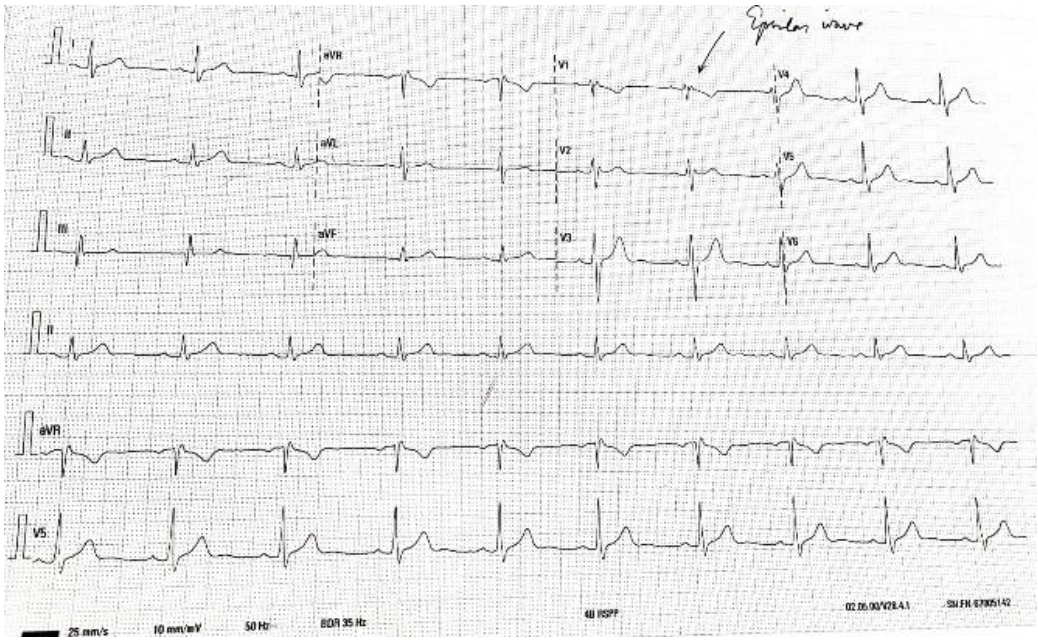

Figure 1. Epsilon waves in the right precordial leads

exhibit any symptoms.[4] In the first phase, known as the "subclinical" stage, there are hidden alterations in both function and structure. However, it is important to note that SCD might still present as the first symptom during this stage. The second phase is characterized by "overt electrical" manifestations, including the presence of RV arrhythmias and the identification of both structural as well as functional abnormality seen in ECG examination. The third stage is characterized by "right-ventricle dysfunction" when there is significant impairment of the right ventricle, while the left ventricle remains unaffected. The fourth stage, known as "biventricular or late," is characterized by substantial impairment of both the RV and left ventricle (LV).[5] ARVC is known to have a strong association with physical activity and may manifest at an early stage, with more severe symptoms seen in individuals who participate in competitive sports. A research conducted by Ruwald et al. found that performing competitive sports increases the incidence of ventricular tachyarrhythmias and SCD by two times compared to participating in recreational sport.[6] Thus, ARVC has been identified as the cause of sudden unexplained death in 11-22% of young athletes.[7]

Making an accurate diagnosis of ARVC is challenging and constantly improving. The criteria have been revised in 1994, 2010, and most recently in 2020, known as the 2020 International criteria or "Padua criteria," which are established by combining several diagnostic criteria. A definite diagnosis of ARVC requires two major, one major with two minor, or four minor criteria from different categories; a borderline diagnosis requires one major with one minor, or three minor criteria, and a possible diagnosis requires one major or two minor criteria from different categories. The primary novelty of the 2020 International criteria was the use of LGE to CMRI tissue characterization data for the identification of fibrofatty) myocardial replacement in both ventricles, which serves as the foundational diagnosis in our case. [8]

Nearly 90% of individuals diagnosed with ARVC have an abnormality on the ECG. Prolonged S-wave upstroke > 55 ms in V1–V3 (90%–95%) is one of the most prevalent findings, and in half of patients presenting with VT, it is followed by an inverted T wave on precordial leads (V1–V3). Furthermore, 30–33% of individuals with ARVC may have an epsilon wave, or depolarization anomaly, which is characterized as a unique wave at the end of the QRS complex.[4] A Holter monitor may reveal non-sustained ventricular tachycardia (NSVT) with varying morphologies. Research using 24-hour Holter monitoring has shown that the majority of patients with ARVC have numerous premature ventricular contractions (PVCs), either occurring isolated or in coupled, exceeding 500 PVCs during 24 hours. Various studies have shown that these premature PVCs and NSVTs are associated with an increased risk of ventricular arrhythmias (VA) and SCD. [9] Moreover, EPS may be used as a diagnostic tool to identify for inducible VT. EPS is a substantial diagnostic test used to differentiate between ARVC and idiopathic right ventricular outflow tract tachycardia. EPS may also give significant information for assessing the risk and making treatment decisions in patients with ARVC. [10]The occurrence of NSVT ($p < 0.001$), inducibility at EPS ($p = 0.005$), and a Holter with frequent PVC ($p = 0.024$) were shown to be significant indicators of effective ICD treatment in a retrospective investigation of 84 patients who

received an ICD for primary prevention. [11] Echocardiography can detect localized RV wall motion abnormalities, increased RV dimension (particularly at the RV outflow tract), and decreased RV ejection fraction (EF). With the advent of new technologies, CMRI has played an important role in diagnosing ARVC. CMRI may detect global or localized ventricular dilation, dysfunction, intramyocardial fat, aneurysmatic dilation, and fibrosis.[12]

Our patient's diagnosis of ARVC is confirmed by the presence of one major criterion and two minor criteria. These include the identification of right ventricular dyskinesia and transmural LGE in the apical right ventricle on CMRI, the presence of epsilon waves in the right precordial leads on electrocardiogram ECG, and the occurrence of frequent ventricular extrasystoles (>500 per 24 hours) on 24-hour Holter monitoring.

Patients with ARVC can present with a broad range of symptoms or they may continue to be asymptomatic. Hence, the treatment approach must be tailored to the patient's specific needs, taking into account their clinical symptoms, risk assessment, and the preferences of both the patient and clinician. Individuals diagnosed with ARVC who have encountered a sustained ventricular arrhythmia are at significant risk of having recurrent sustained VT and VF (or VFL, characterized by a cycle duration of 240 milliseconds or less).[13] During an EPS, the inducibility of sustained VT is another indicator of electric instability. Inducibility during EPS has been discovered as a risk marker in individuals with ARVC who have had an ICD according to studies. [9] These supports a recommendation for ICD implantation in our patient, with a spontaneous polymorphic ventricular VT was found regardless of the induction VF in our EPS.

Several studies have also shown that ICD treatment is an effective strategy for both primary and secondary prevention of SCD in individuals with ARVC. Individuals diagnosed with ARVC often have a youthful age and are anticipated to have a long lifespan when using the implanted device and leads. According to research, the death rates for both cardiac and noncardiac causes after implanting an ICD in individuals with ARVC are low. The cardiac mortality rate per year was 0.9%, whereas the noncardiac mortality rate per year was 0.8%. However, ICD treatment has potential risks in individuals with ARVC. One common issue that occurred was difficult ICD lead installation (18.4%). Lead displacement (3.3%), infection (1.4%), and lead malfunction (9.8%), were other issues associated with ICDs.[14]

Conclusion

The first manifestation of ARVC ranges from being without symptoms to SCD. Consequently, timely identification of the condition is crucial for preventing further mortality. Recognizing the potential ECG findings of ARVC is crucial in determining the need for additional investigations and the implementation of necessary therapies. Evaluating clinical, and cardiac imaging characteristics is crucial for diagnosing and determining the risk level of individuals with ARVC. We successfully delivered accurate diagnosis and treatment, therefore preventing the potentially life-threatening consequences. EPS and Holter monitoring may serve as diagnostic tools to assess the need for implantable cardioverter-

defibrillator (ICD) placement as a therapeutic strategy for patients with ARVC.

Conflicts of Interest

The author started there is no conflict of interest

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