

# Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Presenting with Biventricular Heart Failure

*Biventriküler Kalp Yetmezliği ile Karışımına Çıkan Aritmojenik Sağ Ventrikül Kardiyomiyopatisi (ASVK)*

## Abstract

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a disorder which is attracting increased awareness in clinical practice. Despite being an uncommon disease, it is a frequent cause of unexpected death in young persons. Although the term ARVC used for this cardiomyopathy suggests that it involves the muscle of the right ventricle, in recent years there were cases reported in which the left ventricle was severely affected. The current study reports an ARVC-diagnosed fifteen-year-old male patient who had no clinical features previously and died from sudden congestive heart failure; he had two siblings with a history of sudden unexpected death. We would like to bring to mind once again the important role of early diagnosis for ARVC to avoid undesirable consequences of the disease.

**Key Words:** arrhythmogenic right ventricular cardiomyopathy (ARVC); biventricular heart failure; adolescent

## Özet

Aritmojenik sağ ventrikül kardiyomiyopatisi (ASVK), farkındalığı giderek artan bir klinik tablodur. Seyrek görülmesine rağmen gençlerde beklenmedik ölümlerin sık karşılaşılan nedenlerindenidir. Sağ ventrikülün baskın tutulumu nedeniyle aritmojenik sağ ventrikül kardiyomiyopatisi olarak adlandırılmış olmasına rağmen, son yıllarda ciddi bir sol ventrikül tutulumunun eşlik ettiği tablolar da tanımlanmıştır. Bu yazıda, daha öncesinde herhangi bir klinik belirti vermeyen, iki kardeşinde de ani ölüm öyküsü olan, ani konjestif kalp yetmezliği nedeniyle kaybedilen on beş yaşında bir erkek hastayı sunduk. Erken tanı sayesinde istenmeyen sonuçların önüne geçebilme şansımız olduğunu, bir kez daha hatırlatmak istedik.

**Anahtar Kelimeler:** aritmojenik sağ ventrikül kardiyomiyopatisi (ASVK); biventriküler kalp yetmezliği; adölesan

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## INTRODUCTION

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a kind of heart muscle disease characterized by the gradual replacement of the myocardium with fibrous tissue and fat. It could be a major cause of sudden cardiac death. The diagnosis of ARVC is based on the presence of familial inheritance, structural and functional pathologies of the right ventricle, arrhythmias, and depolarization and repolarization abnormalities in the ECG (1). Data about prevalence are variable due to difficulties in diagnosis. With regard to etiology, acquired and inherited causes have been shown. In a large proportion of patients (30–80%) the disease is familial, primarily autosomal dominant with variable penetrance. In addition, two autosomal recessive syndromic forms of ARVC, namely Naxos disease and Carvajal syndrome, have been found to have a 100% genetic penetrance. The dominant form of the disease shows polymorphic expressivity ranging from complete lack of symptoms to severe disease phenotype experiencing sudden death, a variety that has been attributed to modifier genes, environmental factors and gender effects (2). Clinical prognosis is determined by the rate of myocardial mass involvement. Arrhythmias and heart failure in patients lead to progressive cardiac involvement, and prognosis is poor. Annual mortality is reported to be 3%. ARVC is present in 50% of patients with normal physical examination, and some affected patients may be found with the first clinical signs of sudden cardiac arrest (3). Treatment is individually evaluated for each patient. Heart transplantation is recommended as a final therapeutic option in ARVC patients with either severe, unresponsive congestive heart failure or recurrent episodes of ventricular arrhythmias (4). Here is an example of a 15-year-old male patient with a family history of sudden death of two brothers, who was admitted to our emergency department with respiratory distress and heart failure, followed by treatment in the intensive care unit but died despite support.

## CASE

A fifteen-year-old male patient was admitted to our emergency department because of weakness and respiratory distress. There had been no complaint until

three days before admission. His symptoms included malaise, anorexia, nausea, vomiting, and oliguria. Later he experienced also increasing respiratory distress, which prompted his relatives to bring him to the emergency department. The patient had two brothers who were lost due to sudden cardiac death at the age of 8 and 23, respectively.

Physical examination showed that the patient's general condition was poor, and he was somnolent. Oxygen saturation was 98%, heart rate 120/min, blood pressure 127/70 mmHg, respiratory rate 40/min and axillary temperature 36.3 °C. He was in respiratory distress. Respiratory sounds were rough. There were widespread crepitant rales in the basal lung fields, and the heart sounds were getting distant. The abdomen was distended, ascites was present. Capillary refill time was 4 seconds. The patient's ECG revealed common voltage depression in V1 through V3, T wave inversion and epsilon waves (Fig. 1). Cardiothoracic ratio determined by chest x-ray (0.60) was increased. In echocardiography, biventricular dilatation was observed, especially in the right ventricle. The right ventricle was reported to be akinetic. 9 mm pericardial effusion was found behind the left ventricular posterior wall. Left ventricular ejection fraction was 20% and fractional shortening 11% (Fig. 2). Abdominal and chest ultrasound showed wide free fluid in the abdomen and bilateral pleural effusion, especially on the right side of the chest, respectively. The patient was diagnosed with ARVC and congestive heart failure. He was admitted to the intensive care unit where he was connected to a mechanical ventilator, and supportive treatment was started. The patient was then transferred to a tertiary cardiac center for evaluation as a candidate for cardiac transplantation. Although it was determined that the patient was suitable for transplantation, we learned that after a two-week follow-up and treatment he eventually died.

## DISCUSSION

The etiology of ARVC has not been clearly elucidated, the results of fibro-lipomatous infiltration of right ventricular tissue is thought to be hereditary as it is a disease involving structural and functional cardiac abnormalities (1). The diagnosis of ARVC is

based on the structural, histologic, and electrocardiographic findings, observed arrhythmia, and genetic factors as proposed by ARVC task force in 1994 (5). Modified task force criteria were proposed in 2010 (6). The criteria have been modified to incorporate new knowledge and technology to improve diagnostic sensitivity with the important requisite of maintaining diagnostic specificity. In our case, the 15-year-old boy did not have any complaints before. He was brought to our emergency department because of the sudden onset of congestive heart failure. Our patient had signs of right-ventricular depolarization delay and epsilon waves in the electrocardiogram, and his echocardiogram showed severe right-ventricular dilatation and left-ventricular dysfunction, fulfilling the criteria of two major findings. Further, the fact that his two brothers (aged 8 and 23) were lost due to sudden death was considered a minor criterion which was also supportive of our diagnosis (6).

The prognosis of the disease is worsened in proportion to the involvement of myocardial mass. A minority of patients is diagnosed before the age of twenty. Half of the patients have a normal physical examination, and a portion of the first clinical signs may come with sudden cardiac arrest. In ARVC, the most common symptoms are palpitations after a workout, fatigue, chest pain, syncope, and sudden death can occur. The rate of progression varies for each patient. The reasons are thought to be genetic and environmental factors (7). In our case, the patient had not been hospitalized for any cardiac cause previously. The first presentation of the disease was sudden congestive heart failure. With ARVC now included in the category of cardiomyopathies, the former definition of right ventricular global dysfunction and relatively preserved left ventricular function has been challenged recently, and cases with left ventricular involvement have come to be better understood (8). Histopathological studies show that up to 75% of patients have double ventricular involvement (9). Our patient had biventricular heart failure, as evidenced by physical examination findings and echocardiographic examination.

Therapeutic options consist of lifestyle changes, pharmacological treatment, catheter ablation, ICD, and heart transplantation. Pharmacological options in ARVC treatment include antiarrhythmic agents, beta-

blockers, and heart failure drug therapy. Catheter ablation is a therapeutic option for ARVC patients who have VT. Catheter ablation has not been proven to prevent SCD and should not be considered an alternative to ICD therapy in ARVC patients with VT, with the exception of selected cases with a drug-refractory, hemodynamically stable, single-morphology VT (10). Heart transplantation is recommended as a final therapeutic option in ARVC patients with either severe, unresponsive congestive heart failure or recurrent episodes of VT/VF which are refractory to catheter and surgical ablation in experienced centres and/or ICD therapy.

The annual mortality rate for the disease is 3% (11). The risk of sudden death cannot be assessed and definitive treatment guidelines have not been established. Treatment is carried out and assessed for each patient individually depending on the definition of his/her clinical disease. In our case of a patient with ARVC without other symptoms, we would like to emphasize once again that ARVC can lead to death by sudden congestive heart failure.

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