CASE REPORT

An ECG changed the life of a young boy: a case of arrhythmogenic right ventricular dysplasia

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SUMMARY

Arrhythmogenic right ventricular dysplasia (ARVD) is a progressive condition with the right ventricular myocardium being replaced by fibrofatty tissue. It is a hereditary disorder mostly caused by desmosome gene mutations. The prevalence of arrhythmogenic right ventricular cardiomyopathy is about 1/1000–5000. Clinical presentation is usually related to ventricular tachycardias, syncope, presyncope or ventricular fibrillation leading to cardiac arrest, mostly in young people and athletes. We report a case of a 17-year-old boy from Turkey, who was referred to our cardiology department for an ECG, required of him prior to joining a football team.

replacement of the right ventricular (RV) myocardial tissue by fat and fibrosis, constituting the anatomic basis for reentry ventricular arrhythmias. It involves mostly the RV leading to RV dysfunction and less commonly the left ventricle.² Pathological examinations of patients with ARVD has revealed fibrofatty replacement of the myocardium of the RV. The usual clinical presentation of ARVD is that of palpitations, and non-sustained and sustained ventricular arrhythmias. Syncope or sudden cardiac death (SCD) may be the first manifestation of the disease.³ In advanced stages it may lead to right heart failure.³ In this report, we describe a case of a 17-year-old boy, who was referred to our cardiology department for an ECG, required of him prior to joining a football team.

BACKGROUND

Arrhythmogenic right ventricular cardiomyopathy or dysplasia (ARVD), which was first described in 1982 by Marcus *et al*,¹ is an inherited condition characterised by progressive degeneration and

CASE PRESENTATION

A 17-year-old boy referred to our cardiology department for an ECG. From the patient's history, we learnt that he had *syncopal episodes 2 weeks*

Figure 1 ECG of frequent ventricular extrasystoles in left bundle branch block pattern and non-sustained ventricular tachycardia with inferior axis.

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Figure 2 (A) Transthoracic echocardiographic images of the right ventricular (RV) dilation with depressed thinning of the RV free wall. (B) Cardiac MRI of the RV dilation with aneurysmatic ectasia of the RV lateral wall. (C and D) Cardiac MRI: extensive fat infiltration of the enlarged RV free wall during T1-weighted (C) and conventional spin-echo images (D).

ago. Physical examination was normal, except for frequent extrasystoles. ECG at admission showed evidence of sinus rhythm, frequent ventricular extrasystoles in left bundle branch block (LBBB) pattern and non-sustained ventricular tachycardia (VT) with inferior axis (figure 1). Echocardiography showed moderate dilation of RV, with depressed RV systolic function and thinning of the RV free wall. Left ventricular size and function were normal (figure 2A). ECG and 2-dimensional echocardiogram suggested heart disease involving the RV. Cardiac MRI (CMR) was performed showing dilation of RV with aneurysmatic ectasia of the RV lateral wall (figure 2B), resulting in dyskinetic bulges during contraction. High-intensity areas indicated fatty infiltration of the RV lateral wall during T1-weighted (figure 2C) conventional spin-echo images (figure 2D). The diagnosis of ARVD was made based on ECG features and structural abnormalities seen on imaging. VT was induced in the electrophysiological study. The patient underwent placement of an implantable cardioverter defibrillator (ICD).

INVESTIGATIONS

The genes responsible for ARVD may follow two forms of inheritance, autosomal dominant (AD) and autosomal recessive. The AD form is the most common. Thirty per cent cases of ARVD occur within families; therefore, after the diagnosis is made in one individual, all of his/her first-degree relatives should be screened. The first-degree relatives of our patient were evaluated with an ECG, signal-averaged ECG, exercise

stress test, 24 h Holter monitor, echocardiogram and/or CMR. Our patient was referred to a center with a genetic laboratory.

DIFFERENTIAL DIAGNOSIS

RV outflow tachycardia is a benign disorder that may mimic ARVD owing to the occurrence of exercise-induced LBBB morphology VT. The heart is structurally normal, and there is no history of ARVD or SCD in the family. ECG in RV tachycardia shows upright T waves in V2–V5 and epsilon waves are absent.

TREATMENT

Currently, no definitive treatment is available to cure the disease. The aim of therapy is to prevent death from VT and SCD. This is effectively accomplished by using an ICD. If a life-threatening arrhythmia is detected, the device ‘shocks’ the heart back to normal rhythm. Milder forms of the disease with no symptoms suggestive of arrhythmia may be treated with medications such as β -blocking agents, for example, metoprolol.

DISCUSSION

ARVD is characterised by LBBB pattern ventricular arrhythmias. Morphologically, it is characterised by RV myocardial infiltration with fibrofatty tissue. The infiltration usually occurs in the diaphragmatic, apical and infundibular region known as the ‘dysplasia triangle’, and may develop into dilation or aneurysm. Patients with ARVD may present symptoms at any age, more commonly at a young age, with predominance for male gender

(ratio 3:1).⁴ Palpitations, fatigue and syncope are the most common symptoms. In these patients, SCD due to ventricular arrhythmias can be seen. Estimated yearly mortality rate is 3% if not treated. Sudden death mechanism is the degeneration of VT to ventricular fibrillation.⁵

Fibrofatty islands generate macro-reentries, thus forming the arrhythmogenic substrate. Arrhythmia is induced by adrenergic stimulation as catecholamine infusion or physical activity.⁶ ARVD may cause RV or biventricular heart failure due to dilation and loss of contractile function. MRI allows a morpho-functional analysis of ventricles and identifies adipose tissue. Some authors have suggested that CMR should be used to replace biopsy. The end points that may be identified by CMR are:

Learning points

- ▶ Arrhythmogenic right ventricular dysplasia, a cause of malignant ventricular arrhythmias among apparently healthy young individual, is part of the group of cardiomyopathies.
- ▶ Physicians should consider this condition especially in young individuals presenting syncope and palpitation.
- ▶ After the diagnosis is made in one individual, all of his/her first-degree relatives should be screened.

fibrofatty infiltration in RV with hyperintense signals, fibrofatty infiltration causing diffuse reduction of myocardial thickening, aneurysm or dilation of RV and of RV exit pathway, segmental contraction and global systolic or diastolic dysfunction.⁷

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Competing interests None.

Patient consent Obtained.

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