Magnetic Resonance Imaging Presentation of Arrhythmogenic Right Ventricular Dysplasia: a case report

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Arrhythmogenic right ventricular dysplasia (ARVD) is a nonischemic cardiomyopathy involving primarily the right ventricle. Its characteristics include hypokinetic areas involving the free wall of the right ventricle, fibrofatty replacement of the right ventricular myocardium, and associated arrhythmia originating in the right ventricle. Genetically, it is a cardiomyopathy with autosomal dominant inheritance, variable expressivity, and incomplete penetrance [1]. The location of the gene was mapped to 14q23-24 recently [1]. The incidence and frequency of this disease are still uncertain till now. Clinically, it is difficult to diagnose ARVD directly and accurately. Here, we report a case of 17 years old female with ARVD; the magnetic resonance imaging (MRI) revealed severe global dilatation of the right ventricle, and diffuse thinning of right ventricular myocardium with transmural replacement by fatty tissue and we conclude that MRI may play an important role in diagnosis of the disease.

CASE REPORT

This 17 years old girl denied having any systemic disease before. This time, she suffered from persistent vomiting and dyspnea. She ever visited local medical doctor for help where intravenous infusion of some medicines was given. She felt better and was discharged. However, the above symptoms occurred intermittently, thus she was brought to our cardiovascular outpatient department for help. Besides, she denied fever, chills, abdominal pain, chest pain, chest tightness, palpitation and orthopnea. According to her mother’s statement, one episode of unconsciousness occurred last month.

In the ward, extreme paleness and unstable hemodynamic status (temperature = 36°C, pulse rate = 240 /min, respiratory rate = 20 /min, blood pressure = 94/31 mmHg) were noted; laboratory examinations showed elevated glutamate oxaloacetate transaminase (GOT) / glutamate pyruvate transaminase (GPT) (1539/1329 U/L), and EKG showed left bundle branch block-type ventricular tachycardia. According to her mother’s statement, one episode of unconsciousness occurred last month.

The chest x-ray film (Fig. 1) revealed severe cardiomegaly, and echocardiography showed marked
right ventricular enlargement. Magnetic Resonance (MR) images of heart (Fig. 2a, 2b) were obtained in a superconducting 1.5 Tesla unit scanner (Magnetom Symphony, Siemens, Erlangen, Germany) with body array and spine coil. The result revealed severe dilatation of the right ventricle (RV) and diffuse thinning of right ventricular myocardium. On the T1-weighted dark blood (800/43 [TR/TE]) images, the right ventricular myocardium revealed bright signal intensity; and on the T1-weighted dark blood with fat suppression images (800/43 [TR/TE]), the bright signal intensity seen on the previous T1-weighted images showed decreased signal intensity. Therefore, we got the impression of transmural replacement of right ventricular myocardium by fatty tissue. On the basis of her clinical history, chest films, echocardiography, MR imaging and EKG findings, the diagnosis of arrhythmogenic right ventricular dysplasia (ARVD) was made. Thereafter, she received heart transplantation, and pathologic report of her right ventricle (Fig. 4) revealed replacement of myocytes by adipose and fibrous tissue that is characteristic of ARVD.

**DISCUSSION**

Arrhythmogenic right ventricular dysplasia (ARVD) was first described by Fontaine et al. in 1984. It is a cardiomyopathy with autosomal dominant inheritance, variable expressivity, and incomplete penetrance [1]. The location of the gene was mapped to 14q23-24 recently [1]. The term ‘arrhythmogenic’ is due to the fact that the residual myocytes in the right ventricle set up a re-entry circuit, which leads to tachycardia. More than 35% patients of ARVD are familial cases [2]. It mainly affects young males, and often present with sudden death. The annual death rate of ARVD is around 3%, and it may be reduced to 1% by appropriate medial therapy [3].

The natural history of ARVD mainly involves 4 stages and as followings [3]:

Concealed phase: subtle right ventricular structural changes, with or without minor ventricular arrhythmia, during which sudden death may occasionally be the first manifestation of the disease, particularly in young people with intense physical exertion.

Overt arrhythmic phase: symptomatic right ventricular arrhythmia, and may lead to cardiac arrest and clearly discernible right ventricular functional and structural abnormalities.

Progression and extension of the muscle disease phase: global right ventricular dysfunction, with relatively preserved left ventricular function.

Final stage: when the left ventricle also becomes involved, biventricular pump failure occurs, associated with congestive heart failure, atrial fibrillation, and thromboembolism.

The spectrum of the disease may be separate into fatty and fibrofatty ARVD. Pure fatty infiltration without fibrosis is an early stage of ARVD and with evolution to fibrosis later linked to inflammation.

Diagnostic criteria for arrhythmogenic right ventricular dysplasia have been established considering symptoms, ECG changes, ventricular arrhythmias and structural and functional right ventricular abnormalities (Table 1) [4]. The diagnosis of ARVD is based on a combination of major and minor criteria. To make a diagnosis of ARVD requires either 2 major criteria or 1 major and 2 minor criteria or 4 minor criteria [5].

Multiple imaging modalities can be used for detection of the functional and structural alternations in ARVD, such as conventional angiography, echocardiography, computed tomography (CT), or MRI.
Figure 2. a. The T1-weighted dark blood images of magnetic resonance imaging (TE=43ms, TR=800ms, matrix=256*104) revealed severe dilatation of the right ventricle with diffuse thinning of right ventricular myocardium, and also the right ventricular myocardium showed bright signal intensity. b. on T1-weighted dark blood with fat suppression images (TE=43ms, TR=800ms, matrix=256*104), the bright signal intensity seen on the previous T1-weighted dark blood images showed decreased signal intensity, indicating transmural replacement of right ventricular wall by fatty tissue.

Table 1. Diagnostic Criteria for Arrhythmogenic Right Ventricular Cardiomyopathy

<table>
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<th>Major Criteria</th>
<th>Minor Criteria</th>
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<tr>
<td>Global and/or regional dysfunction and structural alterations</td>
<td>Severe dilation and reduction of RV ejection fraction with no (or only mild) LV impairment</td>
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<td>Localized RV aneurysms ( akinetic or dyskinetic areas with diastolic bulging)</td>
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<td>Severe segmental dilation of the RV</td>
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<td>Tissue characterization of walls</td>
<td>Fibrofatty replacement of myocardium on endomyocardial biopsy</td>
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<td>Repolarization abnormalities</td>
<td>Inverted T waves on right precordial leads (V2 and V3) (age&gt;12 yr; in absence of right bundle branch block)</td>
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<tr>
<td>Depolarization/conduction abnormalities</td>
<td>Epsilon waves or localized prolongation (&gt;110 ms) of the QRS complex in right precordial leads (V1-V3)</td>
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<tr>
<td>Arrhythmias</td>
<td>Frequent ventricular extrasystoles (&gt;1,000/24 hr) (Holter)</td>
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<tr>
<td>Family history</td>
<td>Familial disease confirmed at necropsy or surgery</td>
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It is difficult to use angiography to depict the fibrofatty replacement of the right ventricular myocardium and right ventricular volume change, so angiography is not an appropriate modality for detecting ARVD.

Echocardiography may play a role in the evaluation of patients with suspected ARVD, but it has many limitations in the evaluation of right ventricular function, since there is no generally recommended method to quantify the function of the right ventricle till now. It also does not have the ability of three-dimensional assessment of the heart, spatial resolution, and unlimited field of view that MR can offer.

Kimura et al [6] stated characteristics of the helical CT in the diagnosis of the ARVD. These are the dilatation of the right ventricle, fatty tissue in conspicuous trabeculae of the right ventricle, especially in the anterior wall, apex, and inferior wall, and a scalloped appearance (bulging) of the right ventricular wall. The fatty tissue of the left ventricle and ventricular septum is frequently seen in ARVD, and fat in ventricular septum is another useful finding for the diagnosis of ARVD. Electron-beam tomography and multislice CT are superior to non-gated helical CT in assessment of abnormal right ventricular functions.

Regarding to the MRI about the ARVD changes, the typical presentation [7] that can be demonstrated by MR imaging are as follows: (Reviewing of this patient, the criteria (a) and (b) are matched)
(a) fatty infiltration of the right ventricular myocardium with high signal intensity on T1-weighted images & low signal intensity on T1 fat suppression-weighted images (major criterion)
(b) fibrofatty replacement, which leads to diffuse thinning of the right ventricular myocardium (major criterion)
(c) aneurysms of the right ventricle and right ventricular outflow tract (major criterion)
(d) dilatation of the right ventricle and right ventricular outflow tract (when severe, major criterion; when mild, minor criterion)
(c) regional contraction abnormalities (minor criterion);
(f) global systolic dysfunction (major criterion) and global diastolic dysfunction (minor criterion).

MR imaging [8] also allows a three-dimensional assessment of the heart, the ventricular volume and anatomy; besides, it has excellent spatial resolution and unlimited field of view. It is powerful for detecting not only the fibrofatty replacement of the right ventricular myocardium but also the global regional functional abnormalities of the right ventricular outflow tract and right ventricle. MR imaging can provide the most important morphological, anatomic, and functional criteria for diagnosis of ARVD, such as: cardiac cine can show functional information like regional wall motion abnormality, ejection fraction of right ventricle, and volume of right ventricle, and delayed enhanced IR-prepped gradient echo imaging can show the degree of myocardial fibrosis, which of them cannot be easily provided by CT scan.

Therefore, positive MR imaging findings should be used as important tool in the clinical diagnosis of ARVD, and it appears to be the optimal imaging technique for lesion detecting and follow-up in patients with clinical suspicion of ARVD.

In summary, multiple clinical diagnostic criteria are involved in the diagnosis of ARVD, and MI imaging may play an important and powerful role to identify this disease.

REFERENCES
2. Fletcher A, Ho SY, McCarthy KP & Sheppard MN. Spectrum of pathologic changes in both ventricles of patient dying suddenly with arrhythmogenic right ventricular dysplasia. Relation of changes to age. Histopathology 2006; 48: 445-452
心律失常性右心室發育不良心肌症在磁振造影影像上的表現

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心律失常性右心室發育不良心肌症是一種主要影響右心室的非缺血性心肌病變。其主要特徵包括右心室壁收縮力低下，右心室壁之心肌層被纖維結締組織或脂肪組織取代，及右心室之心律不整。

心律失常性右心室發育不良心肌症是一種顯性遺傳的心肌病變，其表現非常多樣化，近來已發現其病變基因位置為 14q23-24。

本文報告一例有心律失常性右心室發育不良心肌症 17 歲女性病患，其磁振造影影像上的表現為右心室明顯肥大，右心室壁之心肌層廣泛性變薄並且被纖維結締組織或脂肪組織取代。本文旨在描述磁振造影影像可在此類病例中扮演一有用之輔助角色。