Arrhythmogenic Right Ventricular Dysplasia Associated with Bicuspid Aortic Valve

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ABSTRACT

A 22-year old boy was diagnosed arrhythmogenic right ventricular dysplasia (ARVD) with bicuspid aortic valve (BAV) after a symptomatic event of ventricular tachycardia originating from the right ventricle. Cardiac magnetic resonance imaging disclosed right ventricular enlargement, diffuse thinning of lateral wall and normal left ventricle. Echocardiography showed globally thinned, moderately enlarged right ventricle and bicuspid aortic valve. Cardiovascular malformations may be present with bicuspid aortic valve. Here we would like to report the first ARVD case with an accompanying BAV.

Key Words: Arrhythmogenic Right Ventricular Dysplasia, Bicuspid Aortic Valve.

ÖZET

Aritmojenik Sağ Ventrikül Displazisi ile Birlikte Olan Biküspit Aort Kapak

Sağ ventrikül kaynaklı ventriküler taşikardiye bağlı semptomu olan 22 yaşında erkek bir hastada aritmojenik sağ ventrikül displaziye (ASVD) eşlik eden bikuspit aortik kapak (BAK) saptadık. Kardiyak manyetik rezonans görüntülemede sağ ventrikül lateral duvarda yaygın incelme ile genişleme ve normal sol ventrikül saptandı. Ekokardiyografide ise sağ ventrikülde orta derece genişleme ile global incelme ve BAK saptadık. Bikuspit aortik kapak diğer kardiyovasküler malformasyon ile birlikte bulunabilmektedir. Biz bu yazımızda ilk defa ASVD'ye eşlik eden BAK olgusunu bildirdik.

Anahtar Kelimeler: Aritmojenik Sağ Ventrikül Displazisi, Bikuspit Aortik Kapak

INTRODUCTION

Arrhythmogenic right ventricular dysplasia is a genetic cardiomyopathy characterized by ventricular arrhythmias and progressive right ventriculer dysfunction (1). The disease is commonly inherited as an autosomal dominant trait with incomplete penetrance although recessive forms have been described. Here we present the first ARVD case with an accompanying BAV in the literature.

CASE REPORT

A 22-year-old man was admitted to our hospital with intermittent palpitations and syncope. Physical examination revealed a blood pressure of 100/60 mmHg, a heart rate of 120 beats/min but no other abnormalities. An electrocardiogram showed nonsustained ventricular tachycardias and T-wave inversions in the precordial leads. An echocardiogram showed that the right side of the heart was moderately dilated with thinned right ventricular free wall in the absence of intracardiac shunts. Left ventricular systolic function was normal. We also suspected BAV associated with mild aortic regurgitation. An transesophageal echocardiogram showed BAV with mild aortic regurgitation from short-axis view. The short-axis view showed typical BAV with the raphe at the 3 o'clock position and commissures at the 5 and 10 o'clock positions (Figure 1). Cardiac magnetic resonance imaging disclosed right ventricular enlargement, diffuse thinning of lateral wall, global hypokinesia and normal left ventricular function (Figure 2).The age and symptoms of the patient were also in comformity with our diagnosis. As the patient has presented with life-threatening arrhythmia, we implanted implantable cardioverter/defibrillator (ICD) to the patient.

DISCUSSION

Arrhythmogenic right ventricular dysplasia is a genetic cardiomyopathy characterized clinically by ventricular arrhythmias and progressive right ventricular (RV) dysfunction (1). The disease is commonly inherited as an autosomal dominant trait with incomplete penetrance although recessive forms have been described (2). The cause of ARVD is not well-known but recent evidence strongly suggested that it was a disease of desmosomal dysfunction (3).

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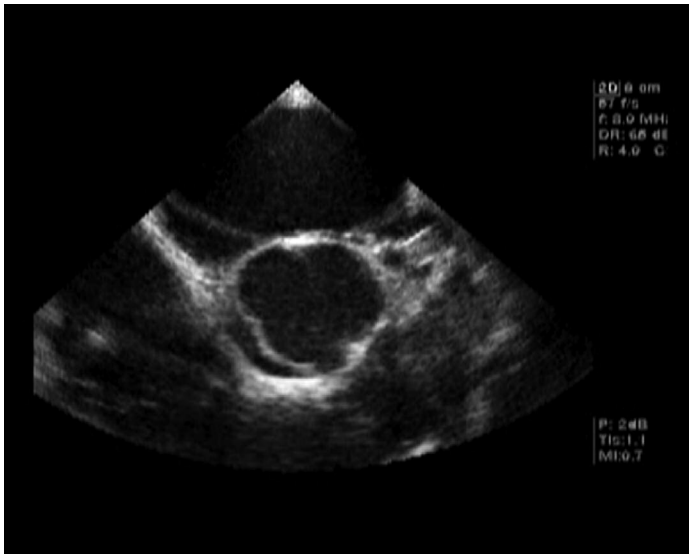

Figure 1: The short-axis TEE view showed typical bicuspid aortic valve

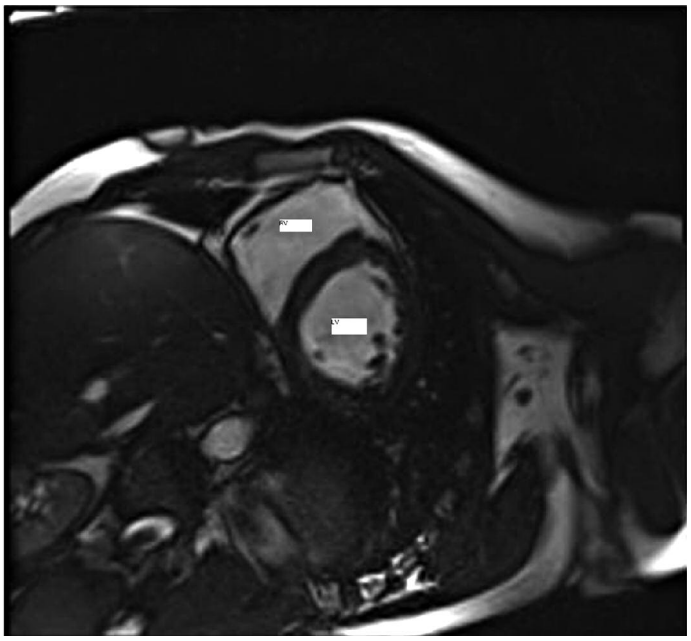

Figure 1: Cardiac magnetic resonance imaging revealed right ventricular enlargement, diffuse thinning of lateral wall, global hypokinesia and normal left ventricular function.

Arrhythmogenic right ventricular dysplasia most commonly presents in young otherwise healthy and highly athletic individuals. The clinical picture may include a subclinical phase without symptoms and ventricular fibrillation being the first presentation, an electrical disorder with palpitations and syncope due to tachyarrhythmias of right ventricular origin or right ventricular or biventricular pump failure. As is typical of ARVD, our patient had familial history and presented with arrhythmia and syncope without congestive heart failure. An electrocardiogram showed nonsustained ventricular tachycardias and T wave inversions in precordial leads. The magnetic resonance imaging showed right ventricular enlargement, diffuse thinning of lateral wall and global hypokinesia. Echocardiogram showed globally thinned, moderately-enlarged right ventricle and normal left ventricular systolic function. Because our patient had ARVD and experienced syncopal episodes highly related to ventricular arrhythmia, the patient had a high risk of sudden cardiac death. We implanted ICD to reduce the risk. The follow-up of the patient was uneventful.

Transesophageal echocardiogram showed that the patient had bicuspid aortic valve with mild aortic regurgitation. Bicuspid aortic valves are the result of an abnormal aortic cusp formation during valvulogenesis. The pathogenesis of congenital aortic valve malformations is unknown. Bicuspid aortic valve is the most common congenital cardiac malformation affecting 1-2 % of the general population, with a higher prevalence in males (4). Bicuspid aortic valves may occur in association with other cardiovascular malformations, including coarctation of the aorta, interruption of the aorta, ventricular septal defect, patent ductus arteriosus, coronary anatomic variants and other conditions such as Marfan's (5) and Turner's syndromes (6). The co-existence of ARVD and BAV has not been reported before. Here we reported such a case for the first time.

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