Congenital heart disease confounding the diagnosis of arrhythmogenic right ventricular cardiomyopathy

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Introduction

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited cardiomyopathy of predominantly the right ventricle (RV) and a cause of sudden cardiac death.[1] Fifty percent of the cases are familial,[2] with causative gene mutations that primarily affect desmosomes and result in progressive fibro-fatty infiltration of the myocardium.[3] In turn, this may lead to ventricular dysfunction and arrhythmias, sometimes causing sudden cardiac death.[1]

There is a putative role of RV stress on disease progression, supported by the observation that the development and manifestations of ARVC may be modulated by the frequency and intensity of exercise in heterozygous ARVC carriers.[4] Even in healthy subjects, chronic strenuous endurance exercise may result in RV dysfunction with a phenotype that is similar to ARVC.[5]

The early diagnosis of ARVC is important to prevent the severe outcomes of unrecognized disease. The 2010 revised Task Force Criteria (rTFC) (Table 1) addresses this issue by increasing diagnostic sensitivity while remaining highly specific.[6] However, it continues to have limitations, especially in children, where some phenotypic features of ARVC may not yet be expressed.[7] Furthermore, the differentiation between ARVC and congenital heart disease (CHD) in the pediatric population is important, as treatments and outcomes vary.

We present a series of pediatric patients in whom the clinical presentation has confounded the diagnosis of CHD versus ARVC.

Case report

We reviewed all cases of suspected or confirmed ARVC between the years 1990 and 2012 at 2 tertiary pediatric hospitals in urban centers. We included patients in whom the examination yielded a presumptive diagnosis of ARVC and a final diagnosis of CHD, or vice-versa. The medical charts were reviewed and clinical, electrocardiographic, and imaging data extracted. The results were anonymized, tabulated, and evaluated under the rTFC. A descriptive data analysis was performed for each case.

We identified 4 patients in whom the findings of undiagnosed or seemingly unimportant CHD were initially attributed to ARVC (Table 2). The median age at presentation was 12 years. They shared in common certain findings: RV or right ventricular outflow tract (RVOT) dilation was present in all, pulmonary to systemic blood flow (Qp:Qs) was increased in 2, and significant ventricular ectopy was noted in 2. The final diagnosis was most often partial anomalous pulmonary venous circulation (PAPVC). In contrast, we identified 1 patient in whom mild CHD masked the likely diagnosis of ARVC.

Case 1

An 11-year-old male subject was referred for evaluation of palpitations, incomplete right bundle branch block (IRBBB) on electrocardiogram (ECG), and over 1000 premature ventricular contractions (PVCs) on 24-hour Holter monitor. Family history was positive for consanguinity. ECG showed an IRBBB (QRS duration 100 ms), T-wave inversion, and slightly negative and biphasic T waves in lead V2. Signal-averaged ECG (SAECG) showed prolongation of the filtered QRS duration and a duration of high-frequency, low-amplitude signals that approached the upper limit of normal (Z-score 1.8). There were over 2000 PVCs on 24-hour Holter monitoring. Exercise testing and echocardiography were both unremarkable. Cardiac magnetic resonance (CMR) revealed a mildly dilated RV with normal contractility. Genetic testing was negative for disease-causing mutations.

At this point, the patient met 2 minor criteria for ARVC and invasive testing was done. Cardiac catheterization with angiography as well as electrophysiology (EP) study were normal. Endomyocardial biopsy yielded subtle mitochondrial changes thought to be noncontributory and inconsistent with ARVC. On follow-up CMR, the RV was moderately

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KEY TEACHING POINTS

- Arrhythmogenic right ventricular cardiomyopathy (ARVC) presents a diagnostic challenge in the young, where phenotypic features may be absent and findings may overlap with congenital heart disease (CHD).
- Right ventricular (RV) volume overload secondary to left-to-right shunting may produce arrhythmias and structural abnormalities that mimic ARVC, and at times meet diagnostic criteria.
- RV dilation despite normal systolic function and increased pulmonary to systemic blood flow ratio were important clues of a covert left-to-right shunt. Cardiac magnetic resonance is useful in differentiating nonobvious CHD from a myocardial disease such as ARVC.

dilated (right ventricular end-diastolic volume index = 141.0 vs 128.7 mL/m^2) and dyskinetic, fulfilling a major diagnostic criterion of ARVC. However, the upper left pulmonary vein was incidentally found to drain to the innominate vein, a finding that was not detected on initial imaging. Subsequent radionucleotide Qp:Qs scanning demonstrated a 1.41 shunt. Given that the patient was asymptomatic, no intervention for PAPVC was required and he was scheduled for clinical follow-up.

Case 2

A 12-year-old male subject was referred for evaluation of syncope leading to a skull fracture with epidural hemorrhage. Family history was positive for sudden cardiac death in a paternal uncle at 50 years of age. ECG showed a QTc duration of 430–460 ms, while Holter monitor and exercise testing were normal. Echocardiography showed RVOT dilation, meeting a minor criterion of the rTFC and raising the suspicion of ARVC. However, CMR showed a left pulmonary vein draining into the superior vena cava. The patient was managed medically for PAPVC and followed on an annual basis.

Case 3

A 17-year-old male subject was referred for mild chest pain and closure of an atrial septal defect (ASD). Echocardiogram showed a small ostium secundum ASD measuring 0.9 cm with left-to-right shunt, low RV systolic pressure (28 mm Hg), and normal interventricular septal curvature. The RV was mildly dilated with a parasternal long axis view of the RVOT of 36 mm or 18.38 mm/m^2 (Z-score of 3.6). This patient fulfilled a major criterion of the original Task Force Criteria and had features of minor criteria of the rTFC on the basis of structural alterations without wall motion abnormality. However, CMR was negative for ARVC. The patient underwent percutaneous ASD closure, which resulted in interval reduction of RV dilation on follow-up echocardiography (Z-score 2.7).

Case 4

A 6-year-old female subject presented for episodic palpitations lasting 30 minutes. Prior evaluation included an echocardiogram showing an ostium secundum ASD, RV and right atrial dilation, and mild mitral regurgitation. Family history was unremarkable. ECG showed normal sinus rhythm with an IRBBB. SAECG showed a prolonged unfiltered QRS duration. Holter monitoring showed rare PVCs and exercise testing induced PVCs of RVOT origin that were suppressed at higher heart rates. A prior ECG had captured a regular narrow-complex tachycardia with a cycle length of 375 ms. She underwent EP study and was found to have dual atrioventricular-nodal physiology. The symptoms persisted despite slow pathway modification. Subsequent echocardiography demonstrated trace localized pericardial effusions around the RV, a 1 cm ASD with left-to-right shunt, and a dilated RV (Z-score 5.34). She was placed on nadolol and underwent percutaneous device closure of the ASD, both resulting in good symptom control.

On routine follow-up at 7 and 10 years of age, she reported episodic syncope. Echocardiography demonstrated persistent RV dilation. CMR showed a right ventricular end-diastolic volume index of 107 mL/m^2 with wall motion abnormalities and a right ventricular ejection fraction of 43%. The left ventricle had areas of delayed enhancement and myocardial thinning with a mildly reduced ejection fraction. Furthermore, history from collateral confirmed that a maternal grandfather had died of a "heart problem" at 42 years of age.

Despite arrhythmia and CHD requiring EP modification and percutaneous closure, respectively, this patient's progressive findings raised suspicion of ARVC. Indeed, this patient met a "borderline" diagnosis of ARVC by the rTFC. The patient was exercise restricted. Subsequent genetic testing did not identify any disease-causing mutations.

Case 5

A 13-year-old female subject was referred for exertional palpitations and dizziness. At 18 months of age a cardiac murmur prompted an echocardiogram that was reported as normal. Family history was positive for a father with "heart attacks" in his thirties. There was a systolic murmur and widely split S2 on physical examination. Noninvasive testing yielded an IRBBB on ECG, ventricular bigeminy on Holter monitor, and RV dilation on echocardiography (Figure 1). At this point, ARVC was suspected. Exercise testing also showed ventricular bigeminy in rest and recovery with occasional PVCs (Figure 2). SAECG could not be interpreted owing to frequent PVCs. Repeat echocardiogram again showed significant RV dilation with normal function and the incidental finding of a left superior vena cava draining to the coronary sinus. The pulmonary veins were poorly visualized. Although no obvious shunt was identified,

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