Case Report

A familial cluster of idiopathic dilatation of the right atrium—A two-case report

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Summary We report two cases of idiopathic atrial dilatation in two adult siblings, a brother and a sister. The first patient was a 36-year-old man who was referred to our institution for evaluation of atrial fibrillation and syncopes. Transthoracic echocardiography revealed an enlarged right atrium accompanied by a severe tricuspid regurgitation associated with annular dilatation. The diagnosis of idiopathic atrial enlargement was made after all other lesions known to produce it have been excluded. The patient did not consent to the proposed cardiothoracic surgical treatment so he remained on conservative therapy. On electrocardiography, atrial standstill was noted, resulting in the implantation of a VVI cardiac pacemaker one year later. After an asymptomatic period, the patient suffered sudden death at the age of 40 years. His 45-year-old sister suffering from the same symptoms was also referred for examination, also to be diagnosed with idiopathic atrial dilatation and severe tricuspid regurgitation. Idiopathic dilatation of the right atrium, although a rare disorder, should not be forgotten as differential diagnosis for enlarged right atrium. Transthoracic echocardiography is the most commonly used technique and in our cases it was sufficient for establishing the diagnosis.

Introduction

Idiopathic dilatation of the right atrium known as giant right atrial aneurysm, is a rare anomaly of unknown etiology [1], which was described for the first time in 1955 [2]. It can be detected at any time between fetal and adult life. Due to its rarity, it can be confused with other more frequent conditions that involve enlargement of the right atrium, such as Ebstein’s anomaly. Both sporadic and familial occurrence have been noted. We report two patients, a brother and sister who were diagnosed with idiopathic dilatation of the right atrium.

Case report

Case 1

A 36-year-old man was referred for cardiological evaluation in our institution for the first time in 2006. He had a history...
A familial cluster of idiopathic dilatation of the right atrium

died suddenly soon after implantation at the age of 39. This brother had been hospitalized several times for episodes of paroxysmal atrial fibrillation, also a finding of dilatation of both atria is mentioned.

Physical examination on admission revealed arrhythmic action of the heart with a grade II/VI holosystolic murmur along the left sternal border, but was otherwise normal. Laboratory findings were also normal. His medication included methyldigoxin and amiodarone. Electrocardiography (ECG) recorded no atrial activity with irregular narrow QRS complexes at a rate of 71 bpm and telemetric monitoring recorded no asystolic pauses. Chest radiograph showed diffuse cardiomegaly (cardiothoracic ratio 0.59) with redistribution of pulmonary blood flow. Echocardiography demonstrated that the area of the right atrium measured 56 cm² (while normal value is 14–22 cm²), annulus of the tricuspid valve 4.0 cm, the area of the grade 3 tricuspid regurgitation jet was 9.5–15 cm². Diastolic diameter of the tricuspid valve 4.0 cm, the area of the grade 3 tricuspid valve into the right ventricle, no morphological changes of the right atrium revealed normal pressures in pulmonary circulation, right and left heart. Pulmonary hypertension, stenosis of pulmonary valve, and left—right shunt as possible causes of right atrial enlargement and massive tricuspid regurgitation were ruled out along with possible coronary artery disease. No signs typical for Ebstein's anomaly were found: there was no downward displacement of the tricuspid valve into the right ventricle, no morphological changes of tricuspid valves, while the size of the right ventricle was normal. Normal thickness and morphology of the right ventricular wall with absence of akinsesia, segmental hypokinesia, or dyskinetic bulges provided no evidence for Uhl's anomaly or arrhythmogenic right ventricular dysplasia. It should also be mentioned that during telemetric monitoring, no ventricular ectopy originating from right ventricle, and in particular no right ventricular outflow tract tachycardia typical for these two anomalies, were recorded. We concluded that the patient had idiopathic dilatation of the right atrium. He refused any possible cardiothoracic surgical procedure and was discharged with the recommendation of avoiding physical exercise and taking methyldigoxin and acetylsalicylic acid as therapy.

One year later, he was readmitted to hospital because of signs of right-sided cardiac decompensation. One month prior to hospitalization methyldigoxin had been removed from therapy by his general physician due to recorded bradycardia. Physical examination now revealed a palpable liver and crural edema. In ECG, a junctional regular rhythm with narrow QRS complexes and frequency of 41 bpm was recorded accompanied by a lack of any electric atrial activity. A control echocardiograph showed a dilated right atrium now with an area of 65 cm² (Fig. 1). Because of malcoaptation of the tricuspid valve, there was a massive grade 4 regurgitation, along with dilated vena cava inferior and liver veins. Subsequently a permanent VVI pacemaker was implanted and after normal electrostimulation was recorded the patient, he was discharged home in good shape and with medication consisting of warfarin, furosemide, and acetylsalicylic acid.

In July of 2008, the patient showed up for a check up, he had no symptoms, and echocardiographic findings were stable (Table 1). Soon after this visit, the patient suddenly died, no autopsy was performed, so that it remains unclear whether a thromboembolic event, a pacemaker malfunction, or a malignant ventricular tachycardia was the cause of death.

Case 2

In August of 2008, the sister of the first patient also underwent a cardiologic examination. She was 45 years old, with no previous cardiological workup done, but with medical history of frequent palpitations and syncopes. Her heart action was arrhythmic, the second heart sound was physiologically split, and there was a soft grade II/VI holosystolic murmur along the left sternal border. Her chest X-ray showed moderate dilatation of the left atrium (LA) 4.7 cm.

Figure 1 Patient 1: transthoracic echocardiogram (finding from 2007)—enlarged right atrium (RA) 65 cm², moderate dilatation of the left atrium (LA) (4.7 cm).

Table 1 Patient 1: comparison of the changes in the echocardiographic data during observation period.

<table>
<thead>
<tr>
<th></th>
<th>2006</th>
<th>2007</th>
<th>2008</th>
</tr>
</thead>
<tbody>
<tr>
<td>LVIDd</td>
<td>4.4 cm</td>
<td>4.1 cm</td>
<td>4.1 cm</td>
</tr>
<tr>
<td>LVIDs</td>
<td>3.4 cm</td>
<td>2.8 cm</td>
<td>3.8 cm</td>
</tr>
<tr>
<td>RVIdd</td>
<td>3.7 cm</td>
<td>3.9 cm</td>
<td>4.4 cm</td>
</tr>
<tr>
<td>LA</td>
<td>4.3 cm</td>
<td>4.7 cm</td>
<td>4.2 cm</td>
</tr>
<tr>
<td>Ao</td>
<td>2.4 cm</td>
<td>3.0 cm</td>
<td>3.0 cm</td>
</tr>
<tr>
<td>IVSd</td>
<td>1.1 cm</td>
<td>1.0 cm</td>
<td>1.1 cm</td>
</tr>
<tr>
<td>LVPW</td>
<td>1.1 cm</td>
<td>1.0 cm</td>
<td>1.0 cm</td>
</tr>
<tr>
<td>LVEF</td>
<td>60%</td>
<td>65%</td>
<td>51%</td>
</tr>
<tr>
<td>RA (area)</td>
<td>56 cm²</td>
<td>65 cm²</td>
<td>65 cm²</td>
</tr>
</tbody>
</table>

LVIDd, left ventricular internal dimension — diastole; LVIDs, left ventricular internal dimension — systole; RVIdd, right ventricular internal dimension — diastole; LA, left atrium dimension; Ao, aorta; IVSd, interventricular septal thickness at diastole; LVPW, left ventricular posterior wall; LVEF, left ventricular ejection fraction; RA, right atrium.
enlargement of the cardiac silhouette (cardiothoracic ratio 0.62) and ECG recorded atrial fibrillation with ventricular response of 61 bpm. Echocardiography demonstrated an enlarged cavity of the right atrium covering an area of 41 cm² and grade 3 tricuspid regurgitation along the dilated vena cava inferior and hepatic veins, and mildly reduced ejection fraction of the morphologically normal left ventricle (Fig. 2). After this one examination, the patient decided to abort the cardiologic workup. Her last therapy included warfarin.

Discussion

Idiopathic atrial dilatation is a very rare anomaly, and there are only sporadic case reports [3]. There appears to be no gender or race preferences, although there are some reports of familial occurrence [4,5].

Accurate diagnosis is essential for adequate treatment, and it is made by systematic exclusion of other, more frequent anomalies, most importantly Ebstein’s anomaly, which is known to produce right atrial enlargement. Massive dilatation of the right atrium is usually associated with tricuspid annular dilatation and tricuspid regurgitation. The diagnostic mainstays are imaging methods, most importantly echocardiography. There is no doubt that other modalities such as transesophageal echocardiography, cardiac catheterization, and cardiac imaging (computed tomography and magnetic resonance imaging) are also useful for evaluation of the right atrium and for excluding other cardiac abnormalities, but transthoracic echocardiography remains the most commonly used technique and here it was sufficient for diagnosing idiopathic right atrial dilatation.

According to a recent review of the literature from 1955 through 1998 for 60 cases of congenital enlargement of the right atrium [6], many patients (48%) were asymptomatic, others presented with arrhythmia, palpitations, chest pain, shortness of breath, fatigue, and syncope. One frequent consequence is also atrial standstill, which leaves cardiac activation dependent on junctional escape rhythm. If the latter however is unstable, it mandates the implantation of VVI or VVIR mode cardiac pacemaker in combination with anticoagulant therapy, as was the case with our first patient.

As far as treatment is concerned, patients with significant tricuspid regurgitation are considered to require tricuspid annuloplasty. Treatment of asymptomatic patients remains controversial. Since they are at high risk of developing atrial arrhythmia and thrombus formation in the right atrial cavity, especially in the setting of atrial standstill, anticoagulation is necessary. It is not surprising that sudden deaths have been reported in a significant number of cases [3].

In our two cases, there was an obvious familial cluster with severe tricuspid regurgitation. Surgical intervention was contemplated and proposed, but both of them declined. The conservative therapy proved insufficient in the first case, resulting in an untimely and sudden death.

In conclusion, idiopathic dilatation of the right atrium, although a rare disorder, should not be forgotten as differential diagnosis for enlarged right atrium. Diagnosis is made by exclusion of other cardiac abnormalities, and screening of relatives is recommended.

References