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Management of Late-Presenting Congenital Combined Heart Defect – Bicuspid Aortic Valve and Ventricular Septal Aneurysm

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Manuscript Preparation E
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3 Department of Cardiac Surgery, University Hospital "St. Ekaterina", Sofia, Medical University, Sofia, Bulgaria**Corresponding Author:** Violeta V. Groudeva, e-mail: violetagroudeva@gmail.com
Conflict of interest: None declared**Patient:** Female, 68-year-old
Final Diagnosis: Bicuspid aortic valve and ventricular septal aneurysm
Symptoms: Cough
Medication: —
Clinical Procedure: Plastic repair of ascending aorta and interventricular aneurysm
Specialty: Cardiac surgery**Objective:** Rare disease**Background:** Bicuspid aortic valve is the most common congenital heart malformation, encountered in 1–2% of the population, while interventricular septal defect and patent ductus arteriosus are the most common congenital malformations associated with bicuspid aortic valve. Although bicuspid valve can have no clinical manifestation, patients with bicuspid valve are prone to develop vascular abnormalities. Aortic dilatation is the most common of these abnormalities, which in turn can lead to serious complications and often requires surgical treatment. Coexistence of bicuspid aortic valve and interventricular septal aneurysm is very extremely rare.**Case Report:** We present a very rare case of a female patient with combined congenital cardiac pathology. The patient was asymptomatic until age 68 years, and presented with nonspecific persistent cough. The diagnostic work-up for the unexplained cough showed normal function of the bicuspid valve and an ascending aorta aneurysm accompanied with interventricular septal aneurysm. The patient was referred for surgery. The diagnostic work-up and the decision for surgical treatment were thoroughly discussed to determine whether it was a true or a false aneurysm.**Conclusions:** The presented case is an example of late diagnosis of a congenital cardiac defect. The silent evolution and the scarce clinical presentation led to incidental discovery of the pathology, which was fully assessed only by computed tomography. Although echocardiography is essential for discovering heart defects, non-invasive imaging techniques are required for detailed morphological assessment and for planning optimal surgical treatment.**MeSH Keywords:** Aorta • Bicuspid • Heart Aneurysm • Heart Defects, Congenital • Heart Septal Defects, Ventricular • Imaging, Three-Dimensional**Full-text PDF:** <https://www.amjcaserep.com/abstract/index/idArt/919766>

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Background

Bicuspid aortic valve is the most common congenital heart malformation and presents in 1–2% of the population, while inter-ventricular septal defect and patent ductus arteriosus are the most common congenital malformations associated with bicuspid aortic valve [1]. Bicuspid aortic valves are prone to develop vascular abnormalities of the aorta, such as dilatation, coarctation, and dissection. The causes range from cystic degeneration to progressive aortic intimal degeneration. About half of young patients with a functionally intact bicuspid valve have echocardiographic data for aortic dilatation, which on its own increases the risk for dissection by 9-fold [1]. Dilatation of the aorta is an important predictor for dissection and rupture of aneurysms in cases of bicuspid aortic valve, even if the valve has normal function [2].

Membranous interventricular septal aneurysms is a rare entity and its exact prevalence is unknown. It is detected in 0.3% of patients with suspected congenital heart disease and in 19.1% with suspected ventricular septal defect [3]. The combination of the 2 entities – bicuspid aortic valve and membranous septal aneurysm – is extremely rare, with few reported cases in the literature.

Case Report

A physically and socially active 68-year-old woman with arterial hypertension was referred to our clinic for a second opinion regarding a newly diagnosed aneurysm of the ascending aorta and was recommended for surgical treatment. Her complaints were nonspecific but she had had a dry cough for almost 1 year, which she said started after a viral infection. Despite 2 courses of antibiotic treatment for superimposed bacterial infection, the cough appeared periodically and in the last 2 months was constant. It was unclear whether the cough was associated with adverse effects of medication. The patient reported episodic pain in the back and chest, without provoking factors. During the diagnostic work-up, the following findings were noted: Echocardiography revealed bicuspid aortic valve with normal function, without stenosis and insufficiency, moderate dilatation of the right heart chambers, with moderate tricuspid insufficiency without right ventricular dysfunction. Systolic pressure in the right ventricle was 38 mmHg. Due to the unclear clinical condition and data from echocardiography, the patient was referred for computed tomography scan to exclude a possible pulmonary thromboembolism or other pulmonary pathology, and to refine the diagnosis. Cardiac CT was performed with Aquillion One (Toshiba) with a protocol triggered to evaluation of the thoracic aorta, coronary circulation, and pulmonary arteries. Images were analyzed and reconstructions were made with dedicated software on a Vitrea workstation.



Figure 1. Bicuspid aortic valve and fusion of right and left leaflets. A 68-year-old female patient referred with a newly diagnosed aneurysm. CT aortography in axial plane reveals bicuspid aortic valve with fusion of right and left leaflets (blue arrow). No raphe is present (black arrow).

The CT did not show signs of pulmonary embolism. Pulmonary arteries were homogeneously opacified, without filling defects. The coronary arteries had normal anatomy and were without significant stenoses. The bicuspid aortic valve was visualized, showing proper coaptation of the leaflets during the multiphasic series (Figure 1). The ascending aorta showed dilatation with maximal diameter at the level of the pulmonary trunk, measuring 61 mm (Figure 2A, 2B). CT revealed a lobulated aneurysm in the membranous part of the interventricular septum, measuring 27×21×25 mm. The aneurysm was protruding towards the cavity of the right ventricle, reaching the tricuspid valve (Figure 3). The findings were confirmed by invasive coronary angiography, which showed normal coronary arteries without stenoses, and increased pressure of up to 35 mmHg in the right ventricle. Aortography revealed normal function of the aortic valve, without insufficiency, and a maximal systolic gradient of 8 mmHg. There was a 60-mm aneurysm of the ascending aorta. During the ventriculography, a large aneurysm of the membranous part of the interventricular septum was visualized, without presence of shunt. There was normal wall motion of left ventricle and preserved left ventricular function. The patient was referred for surgery. Open heart surgery with extracorporeal circulation was performed. The ascending aorta was replaced, with resection of the aneurysm. The aneurysm in the membranous part of the interventricular septum was visualized and seemed to be causing some obstruction of the outlet of the right ventricle. The aneurysm was repaired using

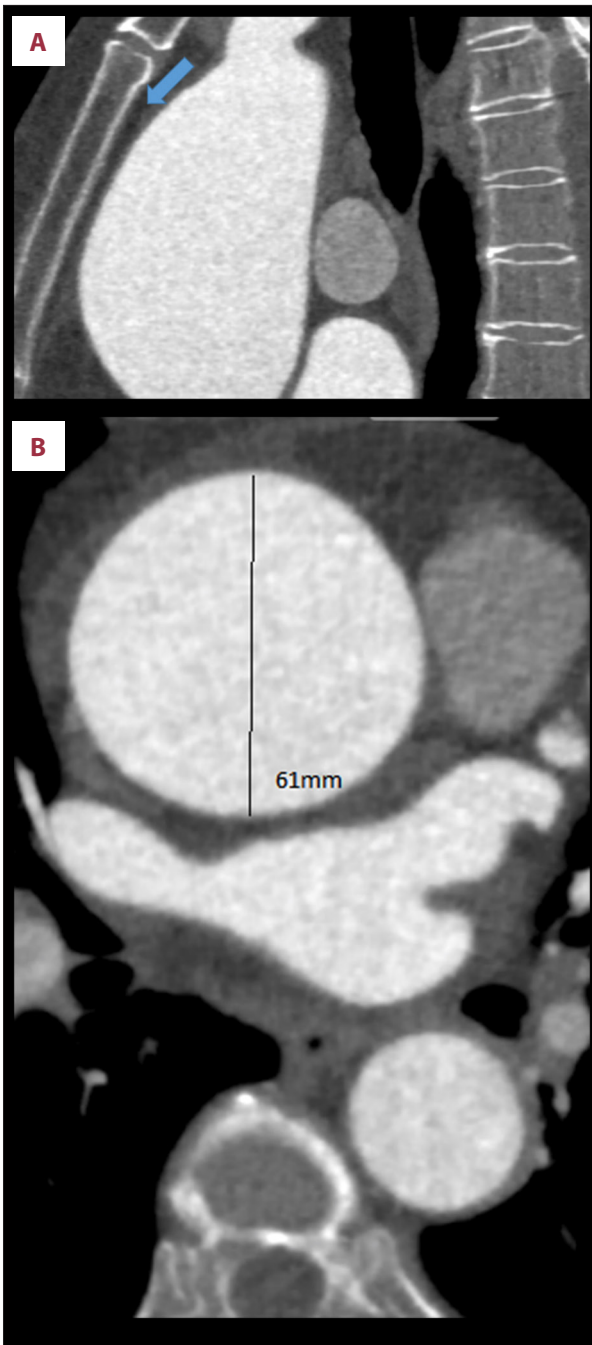


Figure 2. Concomitant aortic aneurysm. Same patient. CT angiography. (A) sagittal plane. Aneurysmal dilatation of the ascending aorta narrowing the retrosternal space (blue arrow). (B) Aneurysmal dilatation of the ascending aorta measured in axial plane at the level of the pulmonary trunk.

a synthetic dacron patch. The postoperative period was uneventful and recovery was normal. Echocardiography showed minimal tricuspid insufficiency, and no signs of pulmonary hypertension were evident. No cough was present. A follow-up

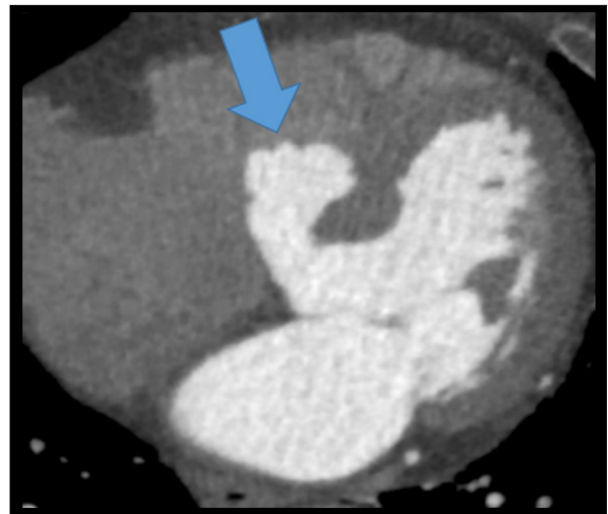


Figure 3. Membranous interventricular septal aneurysm. Same patient. CT angiography, 4-chamber cardiac plane. Membranous interventricular septal aneurysm. Protrusion of the aneurysm into the right ventricle (blue arrow).

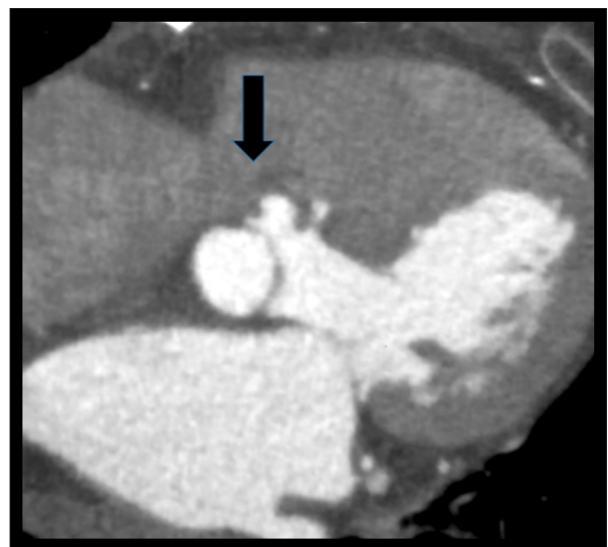


Figure 4. Bulging residual aortic aneurysm. Same patient, follow-up after surgery. CT angiography, axial slice. Residual bulging at the location of the prior aneurysm (black arrow).

CT was performed 7 days after surgery, which revealed normal appearance of the ascending aorta prosthesis. At the site of the previously observed septal aneurysm, there was a slight prominence towards the right ventricle, without forming an aneurysm (Figure 4).

Discussion

The presented case is an example of late diagnosis of evolution of a congenital cardiac defect. Our patient had combined heart defects – a bicuspid aortic valve accompanied with membranous interventricular septal malformation. The patient had no complaints and was asymptomatic until age 68 years. At hospital admission, the clinical and echocardiographic exams showed evolution of the defect, presenting with ascending aorta aneurysmal dilatation and a large lobulated aneurysm of the membranous part of the interventricular septum.

Membranous interventricular septal aneurysm is a rare cardiac anomaly. Its exact prevalence remains unclear, as most of these patients remain undiagnosed. Aneurysm of the membranous septum was first observed by Laennec in 1826 and in 1938 it was defined by Thurman [4]. The aneurysm can be isolated or in combination with another cardiac malformation.

The aneurysm engages the membranous part of the septum and although it is described as an aneurysm, according to Di Cesare, it does appear to be a true aneurysm [5]. Its etiology is unclear. Such aneurysms can develop independently or can develop as a result of spontaneous closure of an interventricular septal defect, as we assume was the situation in the present patient. Some authors divide the aneurysms into true and false aneurysms and pseudo-aneurysms. True aneurysms have a regular shape and broad base, while false ones tend to be irregular in shape and have narrow base. Pseudo-aneurysms are usually secondary as a result of a previous ischemic incident. Studying the anatomical substrate of the saccular prominence of the membranous septum towards the right ventricle cavity, it was demonstrated that the diagnosis “aneurysm of the membranous part of the interventricular septum” is often incorrect. This is because this structure often originates from the tricuspid valve leaflets, not solely from the membranous part of the septum [6].

Nowadays, diagnosis of an aneurysm of the interventricular septum is often an incidental finding during non-invasive exams performed for another reason. It is usually first detected during routine echocardiography exam. According to Canale et al., echocardiography has 57% sensitivity with subcostal 4-chamber view, 62% sensitivity in short-axis view, 71% sensitivity in apical 4-chamber view, and 87.5% sensitivity in long-axis view [7]. Interventricular septal aneurysms are now easily diagnosed by non-invasive methods such as computed tomography and magnetic resonance tomography, which provide detailed visualization of the morphology of the aneurysm. With the constant increase of the number of non-invasive imaging exams performed, interventricular septal aneurysms are more frequently found. They are sometimes discovered during management of complications, usually embolic events [8].

In addition to thromboembolism, complications of IVMS aneurysm can include subacute bacterial endocarditis, left-to-right shunt, obstruction of the right ventricular outflow tract, rupture, thromboembolism, and conduction defects.

Bicuspid aortic valve is one of the most common cardiac pathologies, affecting 1.3% of the population [9]. In such cases, instead of the normal anatomy of aortic valve with 3 cups, only 2 cups are present. It is often accompanied by aortic dilatation and is associated with concomitant cardiovascular pathology. One of the most commonly associated pathologies is dilatation of the ascending aorta. The combination of bicuspid valve and ascending aorta aneurysm was present in our case as well. In bicuspid valves, several morphological appearances are encountered. Usually, 2 leaflets with different sizes are present. Most often, the bicuspid valve is a result of fusion of the left and right leaflets, leading to their anterior–posterior orientation, which is the typical appearance, found in 70–88% of cases. Less frequent is fusion of the right and non-coronary leaflet leading to right-left orientation, which is the atypical form, present in 12–28% of cases. A less common type is fusion of the left and the non-coronary leaflets, found in 0.5–3% of cases [9]. Even less common is the presence of 2 symmetrical leaflets without raphe, as in the present case. The etiology of bicuspid valve is not clearly understood, but is believed to be due to genetic and environmental factors.

Aneurysmal dilatation of the aorta is the most common complication of bicuspid valve. The reported incidence of ascending aorta aneurysmal dilatation in patients with bicuspid valve varies between 20% and 84% [10,11]. The mechanisms leading to aortic dilatation are not clearly understood. Genetic changes in microRNAs are believed to be associated with aortic dilatation in bicuspid valves, although the pathogenesis of the aortopathy is not completely understood [12]. Interestingly, it has been shown that aneurysmal aortic dilatation in the bicuspid valve is not necessarily connected with valve dysfunction, as dilatation can exist in the absence of aortic stenosis or insufficiency [10]. In our case, the bicuspid valve was shown to be by the invasive and non-invasive tests we performed.

The combination of bicuspid valve and membranous interventricular septal aneurysm is extremely rare and few cases are reported in the literature. Deaconu et al. described 2 cases of bicuspid valve and interventricular septal aneurysm [13]; both cases were similar to our patient in that the interventricular septal aneurysm was an incidental finding. One of the cases was a 18-year-old female with prior surgical correction of an aortic coarctation. The patient was a very rare case of bicuspid valve, characterized by fusion of the left coronary and non-coronary leaflets. As the patient was asymptomatic, the aneurysm showed no signs of thrombosis and no ventricular septal defect, and the aortic insufficiency was moderate; therefore,

conservative treatment was proposed with strict follow-up of the valve pathology. The other case had a bicuspid valve with calcinosis and severe stenosis in which the patient presented with symptoms of fatigue, dyspnea, and chest pain. Surgical aortic valve repair and plication of the interventricular aneurysm with tricuspid valve reconstruction were performed.

Our patient was referred to the clinic for surgery due to the incidentally discovered aortic aneurysm. The clinical and echocardiographic exam results helped greatly in fully clarifying the case. The main symptom – unresponsive cough – together with enlarged right heart chambers and moderately increased pulmonary pressure required performing CT to exclude a pulmonary thromboembolism, pulmopathy, or parenchymal mass in the lungs. CT is the criterion standard for assessing these entities. It is debatable whether CT should always be performed in cases with congenital heart defect, even in asymptomatic patients. After discovering the interventricular septal aneurysm, conventional coronary angiography was performed for preoperative assessment of coronary artery status and potential presence of intracardiac shunts. This consecutive approach gave answers to all clinical questions, and the patients was scheduled for surgical treatment. Plastic repair of the ascending aorta was proposed, together with interventricular defect repair. As per ESC guidelines, regardless of etiology, surgery should be performed in patients who have a maximal aortic diameter ≥ 55 mm [14]. The only question that was not fully clarified from the presurgical tests was the nature of the interventricular aneurysm. During surgery, the aortic valve was assessed as competent; therefore, no prosthesis placement was needed. During plication of the membranous part of the interventricular septum, it became evident that the septal leaflet of the tricuspid valve was fused with the wall of the

interventricular septal aneurysm. This case raises questions about the best surgical approach in such patients due to the risk of increasing the tricuspid insufficiency on one hand and the risk for residual aneurysm on the other.

According to some authors, surgical correction of interventricular septal aneurysm also aims to prevent further aneurysm growth and to prevent complications. Cardiosurgical consilium decided to perform combined surgery of the ascending aorta and plication of the interventricular septal aneurysm.

Conclusions

Clinical manifestation and echocardiographic exam are essential for discovering congenital heart defects and their evolution. However, they are not sufficient for complete assessment of the patient's condition. CT should be included in the diagnostic algorithm of congenital heart defects for detailed morphological assessment of the cardiovascular pathology, thus helping surgical planning, irrespective of patient age. Broad discussion is needed regarding optimal surgical treatment in membranous interventricular septal aneurysm fused with the tricuspid valve.

Institution where work was done

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Conflicts of interest

None.

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