Ventricular diverticulum: Definition, pathophysiology, clinical manifestations and treatment

George D. Giannoglou, Stefanos H. Delioglanis, Stelios Paraskevaidis, Yiannis S. Chatzizisis, George E. Parcharidis

1st Cardiology Department, AHEPA University General Hospital, Medical School, Aristotle University of Thessaloniki, Greece

ABSTRACT: Ventricular diverticula are rare abnormalities of the heart, whose origin is not fully understood yet. They are mostly congenital, either isolated or associated with other cardiac and extracardiac defects (Cantrell's pentalogy). Although their etiology is not clear, an embryologic developmental defect has been proposed. Yet, some of them are associated with cardiomyopathies, inflammation or trauma. We discuss the case of a patient with hypertrophic cardiomyopathy and an apical diverticulum. The hypothesis made was that, an obstructing hypertrophic mass, localized in the midportion of the left ventricle, creates a pressure gradient between the apical and basal portions which finally leads to the creation of the diverticulum. Acute rupture, ventricular arrhythmias, peripheral arterial embolism and infective endocarditis are few of their complications. Besides transthoracic 2-D echocardiography and left ventriculography, magnetic resonance imaging (MRI) is the gold standard means of diagnosis. The opinions in the literature, as far as the pharmacological treatment or surgical resection, are controversial.

Key Words: Ventricular diverticulum, Hypertrophic cardiomyopathy.

INTRODUCTION
Ventricular diverticula are very rare abnormalities of the heart, only a few of which have been described in the literature for the past 200 years and whose origin is not fully understood yet. They are mostly congenital, either isolated or associated with other cardiac and extracardiac defects. Yet, some of them are not congenital and only seven cases have been described to associate hypertrophic cardiomyopathy (HCM) with them. We present the case of a man with HCM and a left ventricular apical diverticulum.

CASE REPORT
Our patient, a 69-year-old man was admitted to our hospital with symptoms of dyspnea and tachycardia. Hypertrophic cardiomyopathy (for 30 years), NYHA III heart failure and a DDDR pacemaker (for 2 years due to sick sinus syndrome) were the most important recordings of his medical history.

The physical examination revealed a heart rate of approximately 80 beats/min with a regular heart rhythm while his blood pressure was 105/75 mmHg with 20 breaths/min. An olosystolic murmur 2/6 was heard indicating a possible mitral regurgitation. The pulmonary and abdominal examination was unremarkable. There was a mild edema at lower extremities. The ECG showed an atrial flutter and a pacemaker rhythm. The results of laboratory tests were normal and the chest X-ray revealed a cardiothoracic index of approximately 70%.

The 24-hour Holter ECG did not record anything significant. The Transthoracic Echocardiography (TTE) indicated hypertrophy and dilation of the left ventricle, ejection fraction approximately 50%, mitral and tricuspid regurgitation 2/4, (Right Ventricular Systolic Pressure) RVSP=40 mmHg and, finally, a bulging of the apex of the left ventricle, with thin walls and a narrow neck, which extended over the
right ventricle.

No other significant intracardiac anomalies were found during TTE. The patient was scheduled for an electrophysiologic study and a coronary angiography.

The coronary angiography revealed no atherosclerotic lesions at the arteries and the left ventriculography showed a possible pseudoaneurysm just below the apex (outpouching with a narrow neck and no contractility).

An electrophysiologic study was performed but the attempt for a radiofrequency ablation of the right isthmus had no results. There was a successful electrical cardioversion of the atrial flutter.

The patient was referred to the Thoracic department of our hospital, which proceeded to the resection of the possible pseudoaneurysm. The patient was operated and admitted to the ICU. Thirteen days later he died due to acute renal failure and septic shock. The histological examination of the formation resected, revealed an intense fibrosis with several hypertrophic myocardial fibers and connective tissue. We presume that this was a case of a fibrous diverticulum and its association with HCM is being discussed below.

**DISCUSSION**

Cardiac diverticulum is a very rare malformation of the heart, which is mostly congenital. Since its first description in 1816, 411 cases have been reported, 70% of which involved other extracardiac anomalies while the rest of them were reported as isolated. They
usually arise from the left ventricle but many cases have been presented with right ventricular\textsuperscript{4-7}, biventricular\textsuperscript{8,9} or right atrial\textsuperscript{10} origin.

Although its etiology is not clear, an embryologic developmental defect has been proposed. A failure of normal midline fusion of the paired primitive mesoderm, at the 4\textsuperscript{th} embryonic week, in combination with abnormal fusion of the cardiac loop to the yolk sac before its descent, is believed to result in the development of ventricular diverticula\textsuperscript{11}. Histopathologically, they are classified as fibrous and muscular. As far as the former is concerned, the cardiac wall is constituted mostly of fibrous tissue with few or no muscular fibers. It is non-contractile and most commonly originates from the apical or subvalvular position; it occurs predominantly in black people and Africans. The subvalvular type may be accompanied by aortic or mitral regurgitation and systemic embolism. They are associated with neither defects in the median line nor congenital cardiac malformations\textsuperscript{12}. The muscular type includes all the three layers of the heart and usually emerges from the apex but rarely from the right ventricle or both chambers. It has a mechanical activity, synchronous with the activity of the ventricles. Its connection to the ventricular chamber is narrow\textsuperscript{13,14}. It is frequently associated with other congenital anomalies, including those of the abdominal wall, diaphragm, sternum, pericardium, and the heart itself. Right atrial septal defect\textsuperscript{15}, ventricular septal defect\textsuperscript{16-18}, tetralogy of Fallot\textsuperscript{19-21}, pericardial effusion\textsuperscript{4}, hypoplastic right ventricle\textsuperscript{22}, pulmonary stenosis\textsuperscript{23}, absence of the sternum\textsuperscript{24}, asplenia\textsuperscript{25}, disseminated neonatal hemangiomatosis\textsuperscript{7}, bilateral obstruction of the ureteropelvic junction and agenesis of corpus callosum\textsuperscript{26} are the malformations described in the bibliography. The syndrome, which consists of omphalocele, ventricular diverticulum, anterior diaphragmatic herniation, thoracic ectopia cordis (in its most severe form), along with other cardiac deformities (atrial septal defect, anomalous venous pulmonary return, dextroversion of the heart), is known as the Cantrell’s syndrome\textsuperscript{27-29}. The latter has been identified in many cases involving ventricular diverticula and, even though a cause has yet to be found, genes located on the X-chromosome may be involved\textsuperscript{30}.

However, few cases of diverticula which are not congenital but associated with hypertrophic cardiomyopathy (HCM) have been presented in the literature\textsuperscript{2-3}. The hypothesis made was that an obstructing hypertrophic mass localized in the midportion of the ventricle creates a pressure gradient between the apical and basal portions. The ventricle chronically exposed to high intracavitary pressure gradually changes and a dilated muscular chamber is being created, as the midventricular obstruction worsens. In addition, transmural infarction may occur to HCM due to imbalance between oxygen supply and demand, in the absence of atherosclerotic lesions of the coronary arteries. As a result, subendocardial and apical infarction may eventually create a diverticulum which mostly consists of fibrous tissue\textsuperscript{3}.

Patients with cardiac diverticulum which are not associated with other malformations are usually asymptomatic. ECG abnormalities such as T wave inversion\textsuperscript{31}, ST segment depression and repolarization abnormalities\textsuperscript{32} can be found accidentally during a medical check-up. Chest pain, usually at rest, mostly atypical\textsuperscript{33}, is a symptom that may appear to those patients\textsuperscript{32-34,37}. Supraventricular\textsuperscript{38} or ventricular arrhythmias (ventricular fibrillation\textsuperscript{39}, focal ventricular tachycardia\textsuperscript{40,41}, multiple premature ventricular complexes\textsuperscript{42}) which may lead to syncope or sudden death can be the first clinical manifestation. Patients with diverticula, especially the fibrous ones (because of their non-contractility), may also experience peripheral arterial embolism\textsuperscript{43}. When they are associated with valve impairments, infective endocarditis\textsuperscript{44} may be the first clinical manifestation. Congestive heart failure\textsuperscript{45} and acute rupture\textsuperscript{46,47} have also been described in the literature.

The diverticulum can be easily suspected when it is associated with malformations of the median line as described above. In such a case a pulsatile epigastric mass is very characteristic\textsuperscript{48}. Clinical findings such as heart murmur\textsuperscript{49} and chest pain\textsuperscript{53} or arrhythmias and repolarization abnormalities\textsuperscript{32} found at an ECG must be carefully evaluated. Cardiomegaly\textsuperscript{50} or an abnormal protrusion of the cardiac silhouette\textsuperscript{51} on chest X-ray may provide the earliest clinical suspicion. Transthoracic 2-D and Doppler echocardiography can show the origin, the narrow neck, the walls and the contractility of the diverticula\textsuperscript{52-54}. Left ventriculography\textsuperscript{36,55} with phase analysis can precisely describe the location of the diverticulum, its shape and contractility or other malformations of the heart. Magnetic resonance imaging (MRI) has the poten-
tial not only to identify but also to categorize diverticula non-invasively and differentiate them between muscular contracting and non-contracting fibrous ones.\textsuperscript{56,57} Histological examination, once the resection of the formation is decided, is the one that will verify the synthesis of the diverticulum’s wall and may give information about its cause.\textsuperscript{37,49}

Once the diverticula are associated with other malformations, their resection together with the repairing of the deformities is the most common treatment. Specifically, in Cantrell’s pentalogy, the strategy of repair is divided in two stages: (1) urgent soft tissue coverage and hemodynamic palliation; and (2) intracardiac repair with concomitant chest wall reconstruction and reduction of the heart into the thoracic cavity.\textsuperscript{27} If the patient is asymptomatic and the diverticulum is not associated with other malformations, the opinions, as far as the resection is concerned, are controversial. Because of their life-threatening complications, such as sudden death due to arrhythmias, acute rupture and the possibility of systemic embolization, mostly in the fibrous ones, the surgical resection seems to be the most usual form of treatment seen in the literature. Yet, in asymptomatic patients, many authors suggest long-term anticoagulation therapy,\textsuperscript{43} antiarrhythmic drugs class I and III or beta-adrenergic block agents,\textsuperscript{133} radiofrequency ablation\textsuperscript{33,58} and implantation of cardioverter defibrillators.\textsuperscript{1}

In our case the diverticulum had no contractility and we believed that it was a case of pseudoaneurysm. We decided that the surgical resection was the best treatment for our patient due to symptoms of heart failure (NYHA III) and potential association with systemic thromboembolism and acute rupture, as described in the literature. The histological examination of the resected formation, revealed its true origin afterwards. All diverticula described in the literature which are associated with HCM are fibrous and, as a result, long term anticoagulation therapy or surgical resection seems to be the preferred treatment. The clinical outcome depends on the co-morbidities of the patient.

Περιλήψη: Περιγράφεται σπάνια περίπτωση κοιλιακού εκκόλπωμα (ventricular diverticulum). Ασθενής ηλικίας 69 ετών προσήλθε στο νοσοκομείο με συμπτώματα δύπνοιας και ταχυκαρδιών. Ο ασθενής έπαιρε από υπερτροφική καρδιομυοπάθεια (επί 30 χρόνια), έφερε έντονα καρδιακό βηματοδότη από 2ετίας λόγω σύνδρομου νοσού τον καρδιακό βηματοδότη. Υποβλήθηκε σε καρδιακό καθετηριασμό, ηχωκαρδιογράφημα, ελεκτροφυσιολογική μελέτη και χειρουργική επέμβαση αφαίρεσης εκκόλπωματος. Ο ασθενής παρατηρούσε αύξηση του καρδιοθωρακικού δείκτη, υπετροφία και διάταση αριστερής κοιλίας, κλάσμα εξωθήσεως περίπου 50%, ανεπάρκεια μιτροειδούς και καρδιακογόνως 2/4, προβολή κορυφής αριστερής κοιλίας με λεπτό τοίχωμα και στενό αυχένα. Η αριστερή κοιλιογραφία δεν εδείχθη ανεπάρκεια στις στεφανιαίες αρτηρίες και η αριστερή κοιλιογραφία εδείχθη πιθανό πενδακανεύρισμα αρμόδιοι στον κηρυγματισμένο προβολή της κορυφής αριστερής κοιλίας. Έγινε ελεκτροφυσιολογική μελέτη, αλλά η προσπάθεια για κατάλυση με υψηλότερες πιέσεις έλεγχης είδους ήταν άτοπη. Η προσπάθεια ελεκτρικής ανάταξης της κοιλιακούς μαρμαρίγης ήταν ανεπτυγμένη. Ο ασθενής υποβλήθηκε σε κυτταρική επέμβαση και εισήχθη στη Μονάδα Εντατικής Θεραπείας. Μετά 13 ημέρες ο ασθενής επέβαλα μια φρικτή ανεπαρκεία και στριπτικό shock. Η ιστολογική εξέταση του μυοφύστοματος που αφαιρέθηκε, εδείχθη ανεπάρκεια με πολλές υπερτροφικές μυοκαρδιακές γαρίδες και συνεπεία τον ισορροπιστό ύπνο. Συμπεράνεται ότι η σπάνια περίπτωση εκκόλπωματος της αριστερής κοιλίας μπορεί να εμφανίζεται με τη μορφή δύσπνοιας και αρρυθμιών, σε έδαφος υπερτροφικής καρδιακής καρδιομυοπάθειας.
REFERENCES


