**Sudden death due to ventricular septal defect in a young athlete. A postmortem case report.**

K. Natsis¹, M. Didagelos¹, G. Sofidis¹, S.N. Njau²

¹Department of Anatomy, Medical School, Aristotle University of Thessaloniki, Greece
²Department of Forensic Medicine and Toxicology, Medical School, Aristotle University of Thessaloniki, Greece

**ABSTRACT**

Ventricular septal defect (VSD) is the second most common congenital heart disease and may predispose to sudden death or malignant, life-threatening arrhythmias. We report a case of an otherwise healthy, 20-year-old athlete who died suddenly while sleeping. The autopsy revealed left and right ventricular hypertrophy and a perimembranous VSD with a maximum diameter of 11mm, with no other significant findings on histopathological examination and toxicological analysis.

**Keywords:** ventricular septal defect, congenital anomalies, sudden death, athlete, heart disease

**INTRODUCTION**

Ventricular septal defect (VSD) is the second most common congenital heart disease, after bicuspid aortic valve, having a mean incidence of about 3.57/1,000 live births.¹ It is usually diagnosed, by means of echocardiography, and corrected before adulthood, although spontaneous closure is not rare. Advances in imaging and screening of infants increases the incidence of VSDs to 1.56-53.2/1,000 live births.² Necropsy studies however, present a much lower incidence of about 0.25-1.0/1,000 necropsies, but since the majority of infants with VSD survive, these data do not give a true picture of the condition.³

**Correspondence to:** Konstantinos Natsis, Prof. of Anatomy, Orthopaedic Surgeon, Department of Anatomy, Medical School, Aristotle University of Thessaloniki, Greece
Postal Code: 54124     P.O. Box: 300
Tel/Fax: 0030 2310 999681
Email: natsis@med.auth.gr
There are four major types of VSDs, defined by the location of the defect (Figure 1):4,5

- **Perimembranous/paramembranous/conoventricular**: the most common site of a VSD (80%) is located in the membranous septum
- **Muscular/trabecular**: 15–20%, completely surrounded by muscle
- **Outlet/supracristal/subarterial/subpulmonary/infundibular/supracristal/conal/ doubly committed juxta-arterial**: 5%, located beneath the semilunar valves in the conal or outlet septum
- **Inlet/Atrioventricular (AV) canal/Atrioventricular septal defect (AVSD) type**: inlet of the ventricular septum immediately inferior to the AV valve apparatus, typically occurring in Down syndrome.

Chromosomal, single-gene and polygenic disorders are responsible for these anomalies.2

The aim of our study was to present a case of a fatal VSD in an otherwise healthy young athlete.

**CASE REPORT**

We report the case of a 20-year-old athlete who died suddenly while asleep. He was an active, healthy, symptom free football player without any known family history of sudden cardiac death or other heart disease. He was taking no medication and his family denied any alcohol, drug or dietary supplements abuse. An autopsy was performed in order to investigate the cause of death.

The autopsy revealed a perimembranous VSD (Figure 2) with a maximum diameter of 11mm and a significant degree of left and right ventricular hypertrophy: Interventricular septal end diastolic dimension (IVSd) = 18mm (normal value 6-12mm) and right ventricular free wall = 9mm (normal value <5mm) with a total heart weight of 473gr (normal value 96-200g). The tricuspid, mitral, aortic and pulmonary valves were of normal morphology. The autopsy also showed emphysema and mild lung edema. The histopathological examination of the heart verified the hypertrophy. No other significant autopsy nor histopathological findings were found. The autopsy excluded heart attack, intramural coronary arteries (myocardial bridging) or other congenital malformations. The toxicological analysis did not trace any alcohol or drug abuse.

**FIGURE 1.** Schematic representation of the parts of the ventricular septum (view from the right ventricle). A: membranous septum, B: inlet septum, C: trabecular septum, D: outlet or infundibular septum (B+C+D = muscular septum). TV: tricuspid valve, PV: pulmonary valve.
DISCUSSION

The ventricular septum consists of the muscular septum (further divided in the inlet septum, trabecular septum and outlet or infundibular septum) and the membranous septum (Figure 2). According to this anatomy arise the four types of VSD: perimembranous, inlet, trabecular and infundibular. Perimembranous VSDs include as part of their rim the area of tricuspid-mitral-aortic fibrous continuity (central fibrous body), that may extend to either the inlet, trabecular or infundibular septum and can close by tricuspid valve aneurysm formation. Inlet VSDs are located beneath the septal cusp of the tricuspid valve and possess completely muscular rims. Trabecular VSDs are frequently multiple and are closely related to the edges of the trabecula septomarginalis. Infundibular VSDs are defects with entirely muscular rims located between the ventricular outflow tracts in front of a normally formed and positioned membranous septum and may close by prolapse of the right aortic cusp. However, in general, muscular VSDs can undergo spontaneous closure as a result of muscular occlusion. A reduction in the size of a VSD by any mechanism results in changes in the hemodynamic significance of the defect.

VSDs are usually considered non-life-threatening. However, the clinical presentation and the natural history of patients with a VSD may have many aspects. They may have been operated in childhood with or without residual VSD, there may be insignificant or more severe left to right
shunt with left ventricular volume overload and pulmonary hypertension, or development of Eisenmenger syndrome with shunt reversal (right to left) and cyanosis.5

Nevertheless the presence of a VSD is associated with certain complications, such as a higher risk for endocarditis, heart failure, aortic regurgitation, sub-aortic stenosis, double-chambered right ventricle, complete heart block requiring permanent pacing (in surgically treated patients), severe arrhythmias and sudden death.5,6

Several case reports of sudden death due to VSD have been published.7,8 It is noticeable that most of these patients are of young age, otherwise healthy and participate with full activity, as in our case. The incidence of sudden death in a series of 220 adolescents and young adults with unrepaired perimembranous VSD was about 0.5%, while other studies increase this percentage up to 4.2% with serious arrhythmias (multiform premature ventricular contractions, ventricular couplets, and ventricular tachycardia) occurring in 16-31% of patients, irrespective of VSD size.6,9-11

VSDs may be detected by auscultation, producing a holosystolic murmur. Less frequently they present indirectly on electrocardiography (ECG) from the alterations they gradually pose on the heart (left ventricular volume load and hypertrophy, left atrial enlargement, right axis deviation, right ventricular hypertrophy, right atrial enlargement). Occasionally they present on chest radiography (chamber enlargement, increased pulmonary vascularity, right heart enlargement, dilated main pulmonary artery).2 Nevertheless transthoracic (and when necessary transesophageal and three dimensional) echocardiography remains the gold standard in the diagnosis and evaluation of VSDs (assessment of VSD and valve morphology, hemodynamic evaluation, chamber quantification).2 Magnetic resonance imaging can be helpful in complex lesions and cardiac catheterization can provide accurate hemodynamic measurements and additional morphological data from angiography.2

In our case the presence of the VSD was not known during life, despite its large size. No hemodynamic quantification (mainly by echocardiography) was obtained on the patient. The hypertrophied left and right ventricle, through its large size, should have posed a suspicion. It would have changed cardiac hemodynamics in this young athlete, taking into consideration the increased workload the heart had to compensate for. This young patient, although an athlete, had never undertaken a pre-participation examination, not even an ECG, revealing the problem of inadequate population screening prior to sport involvement.12,13 According to the European Society of Cardiology guidelines, no restrictions are required in patients after a VSD closure, or with small VSDs without pulmonary hypertension, significant arrhythmias, or LV dysfunction, although patients with pulmonary arterial hypertension must limit themselves to low-intensity recreational activity/sports.5

Unfortunately, this is another case of sudden cardiac death in a young athlete, reminding us to reinforce the need for expanding the pre-participation examination in all levels of sport.

REFERENCES
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