Medical Expertise

"Development of the European Network in Orphan Cardiovascular Diseases"
„Rozszerzenie Europejskiej Sieci Współpracy ds Sierocych Chorób Kardiologicznych"

Title: 27-year-old patient with Left Ventricular Noncompaction & impaired systolic function

RCD code: III-5A.1.o

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CASE SUMMARY

27-years-old male, previuosly untreated and physically active, was admitted to the hospital for detailed cardiological evaluation of a new-onset heart failure symptoms. His medical history revealed excessive alcohol abuse and smoking. No other cardiovascular risk factors or history of recent infection were defined.
On admission he was clinically stable. Complained of only mild exercise limitation, class II by NYHA. ECG showed sinus rhythm, signs of atria and ventricular hypertrophy and normal QRS duration. No history of atrial fibrillation was known, nor was it present on the 24-hour ECG Holter monitoring. Holter ECG disclosed 2 episodes of ventricular tachycardia (VT) Elevated levels of NT-proBNP (3371 pg/ml) and liver enzymes were found in biochemical analysis. Echocardiography revealed dilated left ventricle (LV) with severely impaired ejection fraction of 19%, pseudonormal mitral flow with elevated diastolic pressure, moderate mitral and tricuspid valve regurgitation, elevated right ventricular systolic pressure of 47mmHg and preserved right ventricular function. Due to prominent trabuculae of the LV wall seen on the echo study Cardiac Magnetic Resonance (CMR) was scheduled. It confirmed the suspicion of LV non-compaction (LVNC). The authors are hesitant whether this patient should have an ICD implanted and if anticoagulant therapy is advised.

DISCUSSION

LVNC is a rare form of cardiac muscle pathology. It is also known as spongy

myocardium or LV hypertrabeculation. It has been suggested, that LVNC is a primary genetic heart muscle development abnormality, in which the normal, physiological process of cardiac wall development during the intrauterine period is aborted and results in persistence of prominent trabeculae and deep intertrabecular recesses. This spongy myocardium is composed of two layers of compacted and non-compacted muscle [1]. LVNC can be either sporadic or familial with variable inheritance patterns. Autosomal dominant inheritance is more common than X-linked or autosomal recessive [2]. The prevalence of the diseases has not clearly been defined. Studies showed, that it may account for up to 10% of all childhood cardiomyopathies, with a prevalence of 0.12 per 100 000 in the general pediatric population [3]. It was found in 0,014% of adult in echocardiographic registry [4]. Heart failure is the main clinical manifestations of the LVNC (60%–80%). Rhythm abnormalities including atrial and ventricular arrhythmias or sudden cardiac death and systemic cardioembolic events are represent typical complications [2]. Diagnosis is usually established by echocardiography or cardiac MRI [5].

No specific therapy for patients with LVNC has been elaborated so far. The management is targeted at modifying common complications. Society guidelines i.e. ESC Guidelines for the treatment of heart failure are successfully applied and well tolerated by LVNC patients presenting with heart failure. Similarly, indications for the use of implantable devices including ICD or CRT are no different from standard indications in non-ischemic dilated cardiomyopathy. Given the fact that LVNC patients are prone to develop thromboembolic events the use of anticoagulants is widely accepted.

EXPERT'S OPINION

This young patient with LNVC and concomitant impaired left ventricular systolic function and ventricular arrhythmia should be treated according the current ESC Heart Failure Guidelines. The use of betablocker, ACE inhibitor, spironolactone is advised. There are indications for the ICD implantation. Initiation of aticoagulation should be discussed with the patient given the lack of benefit proving trials. He should be advised to quite alcohol abuse.

The patient should be regularly followed – up in the heart failure clinic every 6-12 months with echocardiography and ECG monitoring.

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