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Arrhythmogenic right ventricular cardiomyopathy with multiple thrombi – Case report

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Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited cardiomyopathy commonly transmitted as an autosomal dominant trait, characterized by incomplete penetrance and variable expressivity. So far 13 disease genes have been identified, responsible for approximately 60% of all ARVC cases. The pathological consists of a dystrophy of right ventricle (RV) myocardium with fibro-fatty replacement which leads to RV aneurysms dilatation, providing a supstrate for life-treatening arrhythmias. The course of the disease is divided into four main stages: Subclinical, overt electrical, RV dysfunctional and biventricular late stage. Dilatation of the RV and global or focal wall motion abnormalities are supposed to increase the risk of thrombus formation. It is estimated that annual thromboembolic incidence is 0,5/100 patients, which is extremely rare considering the prevalence of disease being between 1 per 2000 and 1 per 5000 inhabitants. We present the case of 61 year old female patient with multiple thrombi in a cavity of RV who had a combination of ARVC and hereditary trombophilia (homozygous mutation on 5GPAI-1 allele). It is very rare combination and diagnostic challenge for us. Considering that diagnostic protocol went retrograde from rare complication according to cause and that it included anamnesis, laboratory tests, interpretation of ECG and holter ECG monitoring, multimodal imaging methods (TTE, TEE, MRI, multislice CT, PET CT, coronarography), surgical procedure with extirpation of multiple RV masses and pathohistological analysis, and molecular markers of inherited thrombophilia, we believe that this is phenomenal example of the application of personalized medicine into practice.

Biography

Livija Sušić has completed her Faculty of Medicine from J J Strossmayer University, Osijek, Croatia. She is an Internist and Head of Specialist Consultation Department in Health Centre Osijek. She is also a Post-graduate student on J J Strossmayer University, Faculty of Medicine. She is at the beginning of her scientific career and published 4 papers till now.

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