Abstract: 1351


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Topic(s):
Myocardial Disease – Clinical

Citation:

Introduction
A previously healthy 22-year-old Caucasian woman with a history of fatigue and unexplained exertional dyspnea was admitted to our clinic due to palpitations and chest pain. On admission, blood pressure was 110/70 mmHg, ECG showed sinus rhythm 60 bpm with incomplete left bundle branch block configuration. Blood tests showed slightly increased troponin-T 23 ng/L and NT-proBNP 2100 pg/mL.

Procedures
Trans-thoracic echocardiography (TTE) revealed severely enlarged atria, decreased right ventricle function (TAPSE 10mm), restrictive filling pattern and hypertrophy (posterior left ventricular wall thickness 13mm). No wall motion abnormalities were observed and left ventricular systolic function was normal (LVEF 55%). Subsequent cardiac magnetic resonance imaging depicted myocardial oedema on the lateral wall and minor pericardial effusion. Positron emission tomography showed no inflammatory activity. Renewed TTE revealed diastolic flow between the aorta and the pulmonary valve (Figure 1). Multi-detector computed tomography for coronary anatomy analysis showed anomalous origin of the right coronary artery (ARCA) from the left coronary sinus with inter-arterial course between the aorta and pulmonary trunk (Figure 2a & 2b).

Patient management
The patient experienced recurrent chest pain while at hospital with troponin-T peaking at 520 ng/L. Subacute coronary angiography confirmed significant ARCA ostium narrowing (FFR 0.76) while IVUS revealed ARCA compression during systole, suggesting restricted ARCA blood flow. The patient underwent CABG with revascularization of the ARCA territory using right internal mammary artery and after uneventful post-operative clinical course was dismissed from the hospital. Comprehensive cardiology panel gene analysis at follow-up revealed simultaneous heterozygosity for desmin- [DES, c.1360C>T; p.Arg 454Trp] and myotilin-[MYOT, c.1423C>T; p.(Gln475*)] gene variants.

Differential diagnosis
Desmin mutations have been associated with ventricular arrhythmias, atrioventricular conduction disturbances, progressive cardiomyopathy and sudden death. Myotilin mutations are associated with myofibrillar myopathy and peripheral neuropathy with uncertain significance for cardiac disease. In this heterozygous patient for DES- and MYOT- mutations we describe a novel cardiac phenotype of restrictive hypertrophic cardiomyopathy and malignant course ARCA.

Conclusions
Multi-modality imaging and serial clinical assessment may lead to the discovery of rare cardiomyopathy. Genetic testing can offer insight into relations between unique gene profiles and specific cardiac phenotypes and facilitate improved outcomes in such patients. The patient in this case is considered for early ICD-referral due to high risk for sudden death while heart failure follow-up is ongoing due to progressive cardiomyopathy. In such patients genetic counselling and family member screening should be encouraged.

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Figure 1. Echocardiography shows a short axis view at the aortic level where a diastolic flow (arrow) can be seen between the aortic valve (AO) and the pulmonary artery (PA).

Figure 2. Computed tomography with a) contrast and b) volume reconstruction showing anomalous origin of the right coronary artery from the left coronary sinus with inter-arterial course between the aorta and the pulmonary trunk (arrow).