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A not so innocent athlete"s heart

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We report the case of a 17 years old athlete who resorted to the emergency service for palpitations and dizziness during exercising. He mentioned two episodes of syncope associated with exercise in the last 6 months. He was tachycardic (200 bpm) and hypotensive (85/56 mmHg). The electrocardiogram showed regular wide complex tachycardia with left bundle branch block morphology with superior axis restored to sinus rhythm after electrical cardioversion. In sinus rhythm, it showed T-wave inversion in V1-V5. Patient was admitted for study. Transthoracic echocardiography demonstrated mild enlargement and dysfunction of the right ventricle (RV) with global hypocontractility (FAC of 29%). The cardiac magnetic resonance (CMR revealed a RV end-diastolic volume indexed to surface body area of 180 mL/m2, global hypocinesia and RV dyssynchrony, subepicardial late enhancement in the distal septum and in the middle segment of the infero-septal wall. The patient underwent genetic study which showed a mutation in the gene that encodes the desmocolin-2 protein (DSC-2) involved in the pathogenesis of arrhythmogenic right ventricular cardiomyopathy (ARVC). According to the 2010 modified Task Force criteria for this diagnosis, the patient presented 4 major criteria for ARVC (characteristic ventricular tachycardia, repolarization and morphofunctional changes and the presence of pathogenic mutation) and the diagnosis was made. Thus, given the clinical presentation, it was implanted a subcutaneous cardioverter and patient is currently in follow-up at the Cardiology service.

ARVC is present in 1 to 1000-5000 people and is responsible for 20% of all sudden cardiac deaths, especially in athletes. Diagnosis is based on structural, functional, electrophysiological and genetic criteria reflecting underlying histological changes. This case shows and reviews the essential characteristics to the disease recognition and, therefore, to the prevention of its most feared complication: sudden cardiac death.