Characterization of arrhythmic presentation in patients with arrhythmogenic cardiomyopathy

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Background/Introduction: Arrhythmogenic cardiomyopathy (ACM) is one of the most arrhythmogenic conditions known to man. ACM is caused by desmosomal mutations in most cases, resulting in progressive replacement of the myocardium by adipose and fibrous tissue. It comes as no surprise that ACM is one of the leading causes of sudden cardiac death (SCD). Nonetheless, the characteristics of arrhythmic manifestations have not dissected after the release of most recent criteria.

Purpose: This study investigates different types of ventricular tachyarrhythmias which had occurred at first arrhythmic event in patients with ACM.

Methods: We investigated 91 consecutive patients with documented evidence of sustained ventricular tachyarrhythmias from 291 ACM patients followed at our center up to this date. Diagnosis of ACM was made using 2010 Task Force Criteria, and patients were defined as having an advanced disease if they had more than 4 TFC points at diagnosis. Presenting ventricular tachyarrhythmias were divided into (1) life-threatening arrhythmic event (LAE; ventricular fibrillation or hemodynamically unstable polymorphic ventricular tachycardia) and (2) hemodynamically stable monomorphic ventricular tachycardia (MMVT). Right ventricular (RV) involvement was defined as a presence of RV wall motion abnormalities and RV dilation at transthoracic echocardiography or cardiac magnetic resonance.

Results: Our study population was constituted of a predictably higher number of males (n=68; 75%), with an average age at the first arrhythmic event of 38±15 years of age.

At first documented arrhythmia, majority of patients studied experienced a stable MMVT (n=53; 58%), while 38 patients experienced an LAE (n=38; 42%). The patients suffering an LAE as first arrhythmic event were slightly younger than the patients who experienced a stable MMVT (35±14 years vs. 40±15 years; p=0.076) but there were no statistically significant gender differences (28/38 males with LAE vs. 40/53 males with stable MMVT; p= n.s.).

Interestingly, patients who presented with stable MMVT were more likely to have an advanced disease at diagnosis (OR=6.52; 95% CI 2.02–20.99; p=0.002). This is supported by the fact that RV involvement was significantly more common in patients presenting with stable MMVT (OR=4.38; 95% CI 1.26–15.26; p=0.021). Additionally, patients with stable MMVT were more commonly carriers of variants on PKP2 gene (OR=3.6; 95% CI 1.1–11.91; p=0.021).

Conclusions: Our data suggest that two types of arrhythmia reflect the two different stages of the disease. The early stage of the ACM is characterized by LAE in absence of RV structural involvement; while, stable MMVT is typical of PKP2 carriers and advanced stage of ACM with RV involvement.