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Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C): We've come a long way, baby

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ARVD/C was recognized as a clinical entity in 1982 with the referral of patients with ventricular tachycardia who had advanced disease [1]. The diagnosis was straightforward since they had markedly enlarged right ventricles, abnormal right ventricular function, and ventricular tachycardia originating from the right ventricle. The electrocardiograms were strikingly abnormal with the majority of patients having T-wave inversion beyond V2. Subsequently, large-scale studies of this ECG parameter in healthy young and middle-aged population have established that T-wave inversion beyond V1 is present in less than 1% of these healthy young and middle-aged adults [2]. Although anterior precordial T-wave inversion is relatively common in black athletes, they can be distinguished as normal by having ST segment elevation that is rare in patients with ARVC [3]. There was no possibility of genetic confirmation in the original series of patients since the abnormal desmosomal genes had not been discovered at that time. In addition, evaluation by 2D echocardiogram and MRI did not have excellent quality, and imaging interpretation was limited by the few number of patients who were recognized with this disease.

The ability to correctly diagnose ARVD/C took a leap forward with the publication of the revised Task Force Criteria in 2010 [4]. Documentation of desmosomal abnormalities in up to 60% of patients with ARVD/C has also been a major diagnostic advance. Most importantly, it identifies first-degree family members who are at risk [4,5]. This is a two-edged sword, since once a family member is identified as having a desmosomal mutation, he or she may be at risk for ventricular arrhythmias including sudden cardiac death [6,7]. However, due to variability of genotypic–phenotypic relation, the risk of the family member developing the disease is not clear. On the other hand, if a family member of a proband with a desmosomal genetic abnormality does not have the gene, it is highly unlikely that the individual is at risk of developing serious ventricular arrhythmias.

The genetic identification of the first-degree relatives may cause tremendous anxiety in the individual and family due to the knowledge that the disease can cause sudden cardiac death. Therefore, risk stratification is especially important in these individuals. The finding that electrical abnormalities, particularly ventricular arrhythmias such as PVCs or ventricular tachycardia, usually precede structural abnormalities is extremely important for risk stratification in gene-positive family members. It appears that Holter monitoring and

possibly exercise testing to elicit ventricular arrhythmias should be the approach to evaluate asymptomatic relatives rather than testing for structural abnormalities by MRI [8]. In ARVD/C patients with ventricular arrhythmias, insertion of an ICD may be the safest approach to decrease mortality, but ICD implantation over a long period of time is not without problems of inappropriate ICD shocks, infection, lead fractures, etc [9]. Epicardial ablation is a promising treatment [10,11]. It is anticipated that future research will result in identification of molecular and cellular defects in ARVD/C in order to prevent development and/or progression of this disease [12]. The review by Anneline S. te Riele, MD, and Richard Hauer, MD, published in this issue, details how far we have come to verify the diagnosis and improve the treatment of ARVD/C [13].

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