

Clinical diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC) is challenging owing to the broad range of phenotypic manifestations, reduced genetic penetrance and age-related progression characterizing the disease. There is no single “gold-standard” test for ARVC and diagnosis relies on a scoring system of “major” and “minor” criteria based on the demonstration of a combination of defects in ventricular morphology and function, depolarization/repolarization ECG abnormalities, myocardial tissue histological changes, arrhythmias and family history. Definitive diagnosis, based on the Revised 2010 Task Force Criteria (TFC), requires 2 major, 1 major and 2 minor or 4 minor criteria from different categories. Therefore, the initial evaluation of all patients suspected of having ARVC should include physical examination, clinical history, family history of arrhythmias or sudden cardiac death (SCD), ECG, signal-averaged ECG, Holter monitoring and non-invasive imaging tests, typically echocardiography. Following the recent identification of left-dominant and biventricular forms of ARVC, comprehensive imaging of the LV is also indicated.

More recently, cardiac magnetic resonance (CMR) has been introduced in the clinical practice when evaluating a patient with possible ARVC as it can quantitatively assess ventricular volumes and ejection fractions, wall motion abnormalities, and give information on tissue characterization (fatty infiltration and myocardial fibrosis through late gadolinium enhancement). If a non-invasive workup is suggestive but not diagnostic, further testing should be considered including angiography, electroanatomic mapping and rarely endomyocardial biopsy.

The most important task when managing a patient with ARVC is prevention of SCD. Patients with a history of aborted SCD or sustained VT are considered high-risk and should get an ICD. Syncope, non-sustained VT, family history of SCD, severe RV dysfunction, LV involvement and QRS dispersion are considered intermediate risk factors but their individual or combined prognostic value has not been prospectively assessed. Accordingly, ICD implantation should be decided in a case-by-case basis. Radiofrequency ablation should be considered in those patients who are not candidates for an ICD or who have an ICD but get multiple shocks despite pharmacological treatment.

Anti-arrhythmic medications may be used to control symptoms in ARVC. The combination of beta-blockers (sotalol) and amiodarone has been proved beneficial in reducing sustained VT and preventing syncope. Beta-blockers and ACE inhibitors can also be used in ARVC patients particularly those with biventricular dysfunction or heart failure. Cardiac transplantation is indicated in patients with severe heart failure (typically characterizing end-stage disease) and in selected cases with intractable, incessant ventricular arrhythmias.

Particular caution should be addressed to avoid competitive sport activities, which increase disease progression and arrhythmic risk. ARVC is familial in >50% of cases. Screening of family members is therefore of pivotal importance. Clinical evaluation of relatives should be guided by the observation that SCD in ARVC is extremely rare at <10 years of age. Family members who meet TFC should be closely monitored. However, since electrical abnormalities precede structural changes in ARVC, evaluation in family members not meeting TFC should initially focus on the electrical aspects of the disease by ECG and Holter monitoring. Alternatively, genetic testing can be performed and if a pathogenic mutation is identified in the proband, family members can be tested as well to determine whether they are at risk of disease manifestation in the future.