The Genealogy of ARVD - A Review of the Development of Guidelines for the Diagnosis of Arrhythmogenic Right Ventricular Dysplasia

Tessa Sundaram Cook[1], Francisco Contijoch[2], and Saurabh Jha[1]

[1]*Radiology, Hospital of the University of Pennsylvania, Philadelphia, Pennsylvania, United States,* [2]*University of Pennsylvania School of Engineering and Applied Sciences*

Purpose

Arrhythmogenic right ventricular dysplasia (ARVD) is a disease primarily of the right ventricle (RV), with a wide spectrum of presentation that includes sudden cardiac death at a young age. Major and minor criteria for ARVD were set forth in 1994 that included global/regional wall motion abnormalities, endomyocardial biopsy findings, electrophysiologic abnormalities and genetic/family history. The criteria were updated in 2009 to include more specific and quantitative assessments, as well as more detailed requirements for positive family history. Here, we review the specific changes that have taken place and assess the limited evidence available on the impact of the new criteria for the diagnosis of ARVD.

Outline of Content

The 1994 criteria were primarily qualitative, with the exception of a few quantitative cutoffs for electrophysiologic findings. Detection of global/regional dysfunction and structural abnormalities was not predicated on a particular imaging modality, and thus no modality-specific criteria were developed. MRI was shown to play a role in the detection of fibrofatty replacement of the right ventricular myocardium but was felt to underestimate ejection fraction compared to angiography and be potentially problematic in patients with arrhythmia. In the intervening 15 years, the use of MRI for the evaluation of right ventricular structure and function increased in use and acceptance.

Revised task force criteria were developed in 2009. They included specific quantitative cutoffs for identification of structural and functional abnormalities. Specifically, RV outflow tract dimensions on echocardiography and RV end diastolic indices and ejection fractions on MRI were given. On echocardiography and MRI, RV akinesia/dyskinesia alone were no longer sufficient and had to be paired with quantitative evidence of an enlarged, hypofunctioning RV to meet major OR minor criteria. The goal of the revisions was to increase the sensitivity of the guidelines in patients at the less severe end of the disease spectrum as well as family members of probands.

The 1994 task force criteria were found to be highly specific, as the patients used to develop the guidelines all had severe disease. The criteria were not sensitive in patients with less severe disease or family members of the probands. The 2009 criteria showed higher sensitivity (>79% for major criteria and >69% for minor criteria) and high specificity (>95%) when applied to 108 patients enrolled in an ARVD study sponsored by the National Institutes of Health. However, a study of 294 patients in Alberta, Canada referred for evaluation for ARVD showed low sensitivity for the detection of ARVD with the new criteria.

Summary

Task force guidelines for the diagnosis of ARVD have changed significantly since their release in 1994. While the goal of the revision was to increase the sensitivity for detection of less severe disease, more work remains to be done to assess if the new criteria truly improve the diagnosis of ARVD.