Editorial Comment

Familial Occurrence of Right Ventricular Dysplasia: A Study Involving Nine Families*

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Arrhythmogenic right ventricular dysplasia is comparatively new on the cardiologic scene (1). Familial ventricular tachycardia of the left bundle branch type was described by Waynberger et al. (2) in 1974. Marcus et al. (1), in a landmark paper, mentioned that right ventricular dysplasia sometimes recurred in families, and sporadic reports appeared from 1982 to 1987, but now, in this issue of the Journal (3), we have the first major epidemiologic study. Nava et al. (3) used strict criteria that selected only families with cases of sudden death at a relatively young age associated with documented right ventricular dysplasia. Guiraudon et al. (4) classified arrhythmogenic right ventricular dysplasia as a myopathy, and Nava et al. (3) agree that the disease is a primary disorder of right ventricular myocardium and should be included among the cardiomyopathies.

Pathologic findings. At necropsy, the pathology varies from mild focal lesions evident only on histologic study to generalized involvement of the right ventricle with global dilation and widespread areas of segmental dysplasia of the free wall and infundibulum (1,3,5). Left ventricular involvement is morphologically similar although reportedly much less frequent, but its real incidence is unknown (6). Why the left ventricle has thus far not been identified as arrhythmogenic is unclear. The dysplastic myocardium is not necessarily congenitally absent as in Uhl's anomaly (see later), but instead is progressively replaced by fibrous or adipose tissue, or both (3).

Clinical features. The combined observations of Nava et al. (3) and Marcus et al. (1) provide a relatively comprehensive picture of the clinical presentation of arrhythmogenic right ventricular dysplasia. The precise time of onset is difficult to establish, but the disorder occasionally develops in young patients under 2 years of age in whom the initial assessment was normal (3). Asymptomatic right ventricular dysplasia without arrhythmias has been identified in family members of probands. Uther members might have premature ventricular beats of right ventricular origin and recurrent ventricular tachycardia of the left bundle branch type. Sudden death in clinically normal asymptomatic patients is sometimes related to right ventricular dysplasia diagnosed only at necropsy. Conversely, it has recently been argued that accelerated idioventricular rhythm in the right ventricular outflow tract in healthy young patients (7) may represent a benign localized "concealed" variety of arrhythmogenic right ventricular dysplasia (6). Importantly, right ventricular failure is exceptional and physical effort is generally well tolerated despite occasionally marked dilation and a decrease in right ventricular ejection fraction (3). Death due to heat failure occurred in only one patient (3). Pulmonary embolism from the dysplastic right ventricle is relatively rare or perhaps rarely diagnosed (3).

The study of Nava et al. (3) included 72 members of nine families, two of which included three generations. The age range was 8 to 77 years, mean age at death 24 years, male to female ratio 1.5 to 1. In the report of Marcus et al. (1) (confined to adults), the age range was 17 to 65 years, mean age 39 years, male to female ratio 2.7 to 1 (1).

Clinical diagnosis. Although regional right ventricular wall motion abnormalities can be detected by two-dimensional echocardiography in right ventricular dysplasia, right ventricular angiography is currently regarded as the standard for the clinical diagnosis (1,3,5). Defects vary from focal hypokinesia to localized bulges or diverticular outpouchings and deep fissuring (5). Angiographic or echocardiographic abnormalities, or both, are not invariably present, however, and the diagnosis may ultimately depend upon histologic examination of the myocardium (endomyocardial biopsy or necropsy) (5,6).

The scalar electrocardiogram (ECG) only occasionally exhibits a right ventricular conduction defect (1,3) indicating that right ventricular dysplasia does not, as a rule, alter the sequence of ventricular activation. Exercise stress testing may initiate nonsustained ventricular tachycardia and, during intracardiac electrophysiologic study, ventricular tachycardia of the left bundle branch type can usually be induced (1). Two lines of reasoning support the conclusion that the tachycardia is reentrant: 1) late QRS potentials in the scalar ECG imply slow conduction that is a necessary substrate for reentry, and 2) the tachycardia is readily inducible by critically timed stimuli (1).

Given the clinical documentation of right ventricular tachycardia (spontaneous or induced by exercise testing or electrophysiologic study), current information supports the...
following diagnostic strategy to determine whether the cause of the tachycardia is arrhythmogenic right ventricular dysplasia. Two-dimensional echocardiography should be done first, followed by selective right ventriculography if the diagnosis remains in doubt. Endomyocardial biopsy is best reserved for patients in whom the condition is still suspected despite normal noninvasive and invasive studies.

**Treatment.** Treatment focuses on prevention of potentially lethal right ventricular electrical instability. Marcus et al. (1) used antiarrhythmic therapy including type I agents, despite normal noninvasive and invasive studies. These investigators cautioned that, although antiarrhythmic drugs appear to be successful in many patients, the effectiveness of pharmacologic suppression has not been systematically evaluated; nor has it been since that publication.

Surgical attempts at ablation of arrhythmogenic foci have been attempted after failure of antiarrhythmic drug therapy (1). Guiraudon et al. (4) argue, however, that, because the entire right ventricle is potentially arrhythmogenic, single site ablation is likely to be ineffective. When operative intervention is considered necessary, they recommend total disconnection of the right ventricular free wall by surgical exclusion so that there is complete electrical disassociation between the ventricles. This surgical procedure is based on the concept of exclusion described in 1975 as a rationale for surgical management of arrhythmogenic infantile scars. The physiologic role of the right ventricle has aroused interest since 1943. (8) and Guiraudon et al. (4) concerned themselves with the hemodynamic consequences of a procedure that functionally excluded the right ventricle. Experience with atrial dependent pulmonary circulations after the Fontan procedure for single ventricle or tricuspid atresia sheds important and encouraging light on this concern (9).

**References**