

Arrhythmogenic Right Ventricular Cardiomyopathy

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Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an increasingly recognized disorder associated with life-threatening arrhythmias and sudden death in young adults. ARVC may be present in as many as 1/1000 people, although more traditional estimates are from 1/1667 to 1/5000.

In the past, ARVC was most often diagnosed when the heart was examined after the sudden death of individual in their 20's or 30's. Such hearts demonstrated fat and scarring replacing heart muscle in the wall of the right pumping chamber. Increasingly, ARVC is now being diagnosed during life in patients presenting with heart arrhythmias or episodes of fainting, often associated with activity. Because the right pumping chamber has a complex shape, lies directly under the breastbone and is surrounded some normal fat, it is often difficult to see or recognize the small changes that occur in the cavity and wall of this pumping chamber in affected people. Similarly, ECG's in such patients often appear relatively normal unless specific measurements or special kinds of ECG's are performed. ARVC often requires multiple abnormalities to be identified on several tests in order to make a firm diagnosis. Fortunately, the process of diagnosis has been helped by the definition of ARVC Task Force Criteria by an expert consensus committee of the European Society of Cardiology in 1994.

ARVC diagnosis is now moving into an era of potential genetic diagnosis. Multiple gene locations associated with ARVC have been identified. So far, a common feature is that these gene mutations change proteins that hold heart muscle cells together at their ends. Some genes are available for clinical testing through out-of-country laboratories, with funding for this testing assessed by provincial health ministries on a case-by-case basis. Other gene testing is also performed in research laboratories. Currently (2007), genes are only identified for between 15 and 50% of patients with ARVC.

Symptoms and Assessment: The most common symptoms among adults with ARVC are palpitations, syncope and sudden death. Children likely present with similar symptoms, although sudden death is less common. Among patients who died with typical ARVC, only 10% of deaths occurred before age 19, but 50% occurred before age 35. Among children assessed for ARVC, patients rarely met ARVC Task force criteria before age 8 years, and the rare deaths that occurred were after age 12 years.

Electrocardiogram (ECG): ECG features of ARVC in adults, when carefully measured, can be identified in 75 to 84%, and are usually quite specific (not found in unaffected individuals). The Signal-Averaged ECG (SAECG), available as a separated instrument or as an option on many ECG machines, is a method of magnifying and measuring small electrical forces not visible on a regular ECG. These measurements are useful for diagnosis of ARVC, and may help predict if arrhythmias are likely to occur. Signal-averaged ECG parameters in children should be compared to normal values for children, which depend on size.

Exercise Testing: Whether the development of extra beats on exercise testing assists diagnosis in ARVC is unclear. Nevertheless, the risk of serious arrhythmias is increased with certain kinds of exercise, and there is some evidence that exercise may speed up the progression of disease. It is recommended that competitive exercise be avoided, and recreational exercise be restricted according to American Heart Association guidelines.

Magnetic Resonance Imaging (MRI) and Angiograms: Individual MRI findings usually can't make the diagnosis of ARVC, but combinations of major and minor MRI criteria have been developed and are helpful. Major criteria include fatty infiltration the heart muscle and enlargement, aneurysms or reduced function of the right pumping chamber. Newer MRI techniques such as myocardial delayed enhancement (MDE) that can identify scar tissue may be more helpful than previous MRI techniques which attempted to enhance for fat alone. MRI studies with MDE corroborate the finding of significant fibrosis in endomyocardial biopsies from the right ventricular septum. Angiograms, performed in a heart catheterization lab, can also identify abnormalities of the size and function of the right pumping chamber.

Electrophysiologic Studies: Among adults, electrophysiology studies have been used effectively to differentiate ARVC from more benign arrhythmias coming from the right pumping chamber. Measuring and mapping the voltage on the inside of the right pumping chamber can also be helpful in making the diagnosis of ARVC. Catheter ablation is occasionally used in patients with ARVC where the rhythm abnormalities are difficult to control.

Biopsy: Small biopsies taken from the inner wall of the right pumping chamber can assist in the diagnosis of ARVC, sometimes confirming the diagnosis in someone who would not otherwise meet the diagnostic criteria for the disease. This potential benefit must be weighed against the small risk of serious complication rates from biopsy procedures.

Risk stratification:

While just the diagnosis of ARVC is often a challenge, a further assessment of the patient's risk should also be completed. Significant risk factors common to two or more studies include increasing dysfunction or dilatation of the right pumping chamber, dysfunction of the left pumping chamber and spontaneous ventricular tachycardia (VT). Inducibility of VT at EP study may or may not be a risk factor. Increasing abnormalities on SAECG are also associated with poorer prognosis. In asymptomatic individuals with ARVC but no device, frequent assessments with echocardiograms, Holters and SAECG's should be performed to monitor for these risk factors.

Medical Assessment: Given the complexity and implications of the diagnosis of ARVC, individuals with suspected ARVC should likely be assessed by a cardiac electrophysiologist (adult or pediatric) appropriate to their age. One resource to find such medical specialists (by region) is the Heart Rhythm Society: http://www.hrsonline.org/find_heart_rhythm_specialist/default.asp