Title: Arrhythomogenic Right Ventricular Cardiomyopathy *GeneReview* — Noninvasive and Invasive Tests of Cardiac Structure/Rhythm: Additional Considerations
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Noninvasive and Invasive Tests of Cardiac Structure/Rhythm: Additional Considerations

Noninvasive Testing

ECG changes may develop over time; therefore, individuals suspected of having ARVC who do not show these changes should undergo serial ECGs [Quarta et al 2010]. Exercise stress testing may also reveal ECG changes including epsilon waves and ventricular ectopic beats that help identify those at risk [Perrin et al 2013].

Echocardiography

- Echocardiography remains a valuable mode for evaluating the individual with ARVC [Haugaa et al 2016]. Like other imaging modalities, detection of ARVC findings can be subjective and is best performed at centers with expertise in imaging ARVC. Right ventricular mechanical dispersion may be observed in ARVC individuals with ventricular tachycardia [Sarvari et al 2011].
- Echocardiography can be considered for persons with defibrillators for whom cardiac MRI is contraindicated [Prakasa et al 2006, Haugaa et al 2016].

Cardiac MRI

- MRI images demonstrating ARVC are included in Murphy et al [2010].
- Cardiac MRI analysis should also include assessment of global and regional right ventricular function and right ventricular myocardial fibrosis.
- Because identification of right ventricular free wall thinning and fatty infiltration on cardiac MRI in persons with a mild ARVC phenotype can be subject to a high degree of intra-observer variability, cardiac MRI is best performed in centers with experience [Haugaa et al 2016].
- Family members with molecularly confirmed ARVC should be screened and followed in centers familiar with ARVC. For family members of ARVC in which a primary pathogenic variant has not been identified, screening must rely entirely on clinical, rather than genetic, data [Haugaa et al 2016].
- Cardiac MRI has not been able to detect early changes of ARVC in children; however, this could result from the absence of manifestations in children rather than a failure of the method [Fogel et al 2006]. Further studies are needed.

Invasive Testing

- **Electrophysiologic (EP) study findings.** Ventricular tachycardia easily induced with ventricular pacing and extrastimulation
- **Right ventricular angiography (RVA) findings.** Enlarged right ventricle with segmental abnormalities
- **Right ventricular endomyocardial biopsy findings.** Fibrofatty replacement of the myocardium (predominantly in the apex, right ventricular outflow tract, and right ventricular inflow tract) and/or atrophy of the right ventricular myocardium

  Note: (1) The biopsy must sample an affected region to be diagnostic. (2) Studies have demonstrated a role for immunohistochemical staining of the myocardium for intercalated disk proteins [Asimaki et al 2009, van Tintelen & Hauer 2009].

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