Continuing the age of discovery in cardiovascular disease

A rare genetic defect involving the heart muscle is leading to early heart failure and potentially lethal heart rhythms for patients in Saudi Arabia. This disease, called Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia or ARVC/D, involves healthy heart muscle cells being replaced with fatty or fibrous tissue. The fatty/fibrous tissues are not able to pump blood effectively or transmit electrical impulses appropriately and in time this leads to a malfunctioning of the heart including sudden death. This disease was first described in the literature in 1978 but it wasn't until 1994 that the International Task Force Guidelines based on structural, histological, ECG, arrhythmic, and familial features of the disease. This was revised in 2010 including specific quantitative parameters to aid in the definitive diagnosis of the disease.

This study followed 22 patients from 2007 to 2010, who were treated at the Heart Centre, King Faisal Specialist Hospital and Research Centre in Riyadh. The mean age at diagnosis for this group of patients was 33.3 years and the follow up ranged from 29 to 132 months. Most of the patients in the study were men (82%) which is consistent with previous studies demonstrating a higher prevalence of ARVC/D in men. However the male to female ratio was 4.5:1 which is higher than the previously reported ratio of 1.6 to 2.7:1. It is possible that we have fewer female patients due to referral bias and less acceptance of implantable cardioverter defibrillators (ICDs) in our female patients.

Most of our patients presented with palpitations, and or ventricular tachycardia (VT). Previous studies have shown that individuals with ARVC/D present with palpitations, syncope, atypical chest pain or right ventricular failure or are sometimes asymptomatic.

Two of the most important diagnostic tools, electrocardiograms (ECG) and echocardiography, are also two of the least expensive and least invasive. ECG abnormalities were present in up to 90% of ARVC/D patients and the most common abnormalities were T wave inversion, often associated with a slight ST segment elevation, and epsilon waves, found at the end of the QRS complex. Epsilon waves are generally found in up to 30% of ARVC/D cases.

The echocardiogram is also a very helpful and useful method of diagnosis. RV function measurements have played a crucial role in imaging the structural and functional abnormalities of the right ventricle associated with ARCV/D.

Treatment modalities for the 22 patients involved in this study included anti-arrhythmic drugs, radio frequency catheter ablation and implantation of an ICD device.

Drug therapy includes beta blockers, sotalol and amiodarone. Sotalol may be the most efficacious drug but due to the progressive nature of the disease arrhythmias may recur despite initial success of drug therapy.

Radio frequency catheter ablation (RFA) is used in managing patients with ARVC/D who have recurrent ventricular tachycardia. The reported success of ablation varies from 41% to 88% in the acute phase, but chronic success rates range from 7% to 88%. Again, the progressive nature of the disease effects the VT recurrent free survival rates.

Most of our patients have had an ICD implanted and this is considered a standard therapy. ICDs can effectively terminate life threatening arrhythmias in patients with ARVC/D who are considered high risk for sudden cardiac death.

The study of this rare but increasingly recognized heart muscle disease seen in Saudi Arabia and other parts of the world will continue to expand our understanding of the best diagnostic tools and treatment options.

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Publication

Arrhythmogenic right ventricular cardiomyopathy/dysplasia in Saudi Arabia: a single-center experience with long-term follow-up.
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