



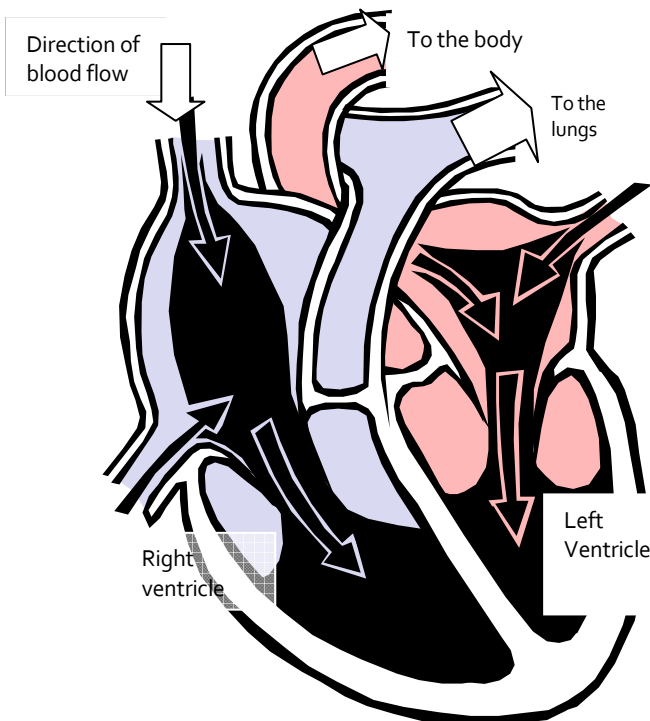
Cardiac Inherited Disease Registry N.Z.

Cardiac Inherited Disease Registry
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Arrhythmogenic Right Ventricular Cardiomyopathy Information

What is Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)?

Arrhythmogenic Right Ventricular Cardiomyopathy or Dysplasia (ARVC or ARVD) may be an inherited heart condition in approximately 30-40% of individuals. ARVC affects the muscle of the right ventricle (one of two main pumping chambers that make up the heart). The right ventricle may become progressively thinner due to muscle cells being replaced by fatty deposits and fibrosis. In some cases the left ventricle may also be affected by ARVC.

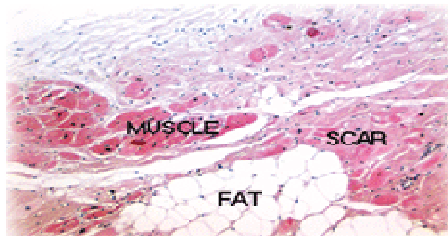


The heart contains 4 chambers, two upper and two lower. The ventricles are lower chambers and are primed (or filled) by two upper chambers at the top of the heart called the atria. The left ventricle fills with blood primed by the left atria, (fully oxygenated from the lungs) and pumps this blood filled with oxygen to the brain and body cells.

The right ventricle pumps blood to the lungs, where wastes are removed (by breathing them out) and fresh oxygen is added to the blood by (breathing in). Oxygen that is required by all body cells is then returned to the heart, and pumped around the body by the left ventricle.

Therefore the heart consists of a 2- phase pump that works together to ensure that as oxygen is removed from the blood by body cells it is rapidly replaced.

The words Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) or (ARVD) Arrhythmogenic Right Ventricular Dysplasia as it was formally known, in simple terms means that the right ventricular



Pathology of an ARVD/C Heart

heart muscle is affected by a condition that can cause the heart to beat very fast and erratically. In order to understand these words it is easier to break them down for instance, 'cardio' is another word which describes the heart and 'myopathy' means a condition or problem related to muscle. The word 'arrhythmogenic' describes how the condition normally presents itself, which is through very fast erratic heart rhythms (cardiac arrhythmia) that usually cause people to feel dizzy or collapse, or cause a cardiac arrest (where the heart muscle stops beating effectively and collapse occurs).

Incidence of ARVC

The true incidence of ARVC in NZ is yet to be determined. In Italy it is thought to affect about 1:5000, and is responsible for 20% of sudden deaths in young adults and 25% of sudden deaths among athletes. Since the condition may be inherited in about 30-50% cases, it is recommended that parents and brothers and sisters and the children of those affected should be screened for the ARVC. ARVC has been found to be an important cause of sudden death in young fit athletes, however males and females of all ages, races and activity levels can carry the condition although it typically affects or causes symptoms in adult young men.

Families with two or more affected individuals have been found in a number of countries, such as North America, Italy, Japan, Africa, and Northern Europe, Australia and New Zealand.

Symptoms

Sometimes people may be completely asymptomatic, that is they have no symptoms at all as often the first symptom may be collapse or sudden death.

Other people may have symptoms of an erratic heartbeat (palpitations or a racing heart beat) occasionally as their only symptom. Patients with ARVC usually present with some type of symptom prior to age 45, but often may present in late puberty or before age 30.

Diagnosis of ARVC

ARVC was first characterised by Dr's Marcus and Fontaine in 1982, following the first description of the condition in 1965. In 1996 ARVC was added to the WHO classification of cardiomyopathies. Since then a number of important diagnostic criteria have been developed that can help doctors to make the diagnosis of ARVC.

There is no single diagnostic test to establish or exclude the diagnosis of ARVC. Currently doctors will utilise many available tests in order to diagnose it. An ECG (electrocardiograph) is a recording of the heart rhythm and this may show some of the signs of (other ECG based testing may included 24 hr Holter recordings an Exercise treadmill tests ETT), and Signal averaged ECGs (SAECGs) are all used to aid diagnosis..

Electrophysiology (EP) studies which can map areas in the heart prone to electrical short circuits that may cause abnormal heart rhythms may also be offered to symptomatic patients.

Magnetic resonance imaging (MRI), is a specialised scan that can provide a superior images of the heart and in particular the right ventricle. Echocardiography, utilises an ultrasound beam to image the heart, and may prove to be useful in some cases. Other studies including right heart angiograms, in which a contrast dye is injected into the right heart as it pumps so that abnormalities of function can be visualised. During the preparation for an angiogram a biopsy of the heart muscle can also be taken, and this also provides Cardiologists with valuable information.

Molecular genetic analyses of genes which encode for the connective proteins which bind the heart together (sarcomere proteins) and help it stretch effectively to beat are beginning to reveal some recognisable changes or mutations in some genes. Currently 7 genes responsible for ARVC have been identified, as well as many more candidate genes that have been targeted for research, so it is likely that more genes will be found. In the future testing an individual for genes that are known to cause ARVC and finding one, may allow other family members to be able to be tested to see if they also carry the same gene, although it is important to stress that many people who carry genes for the condition may never manifest the disease themselves, however they are still able to pass it on to their offspring. Although ARVC can be inherited, doctors remain unsure why some individuals who carry genes for the condition, do not manifest any symptoms throughout their lives, whilst others are affected by the condition at an earlier age.

Further research will answer some of these questions and allow Doctors in the future to better predict who may be at risk of developing the condition.

Diagnosis of ARVC is based predominantly on having at least two of the major accepted diagnostic criteria for the condition or at least one major and 2 minor criteria. Diagnosis of ARVC is a skilled process and therefore it may take several different diagnostic tests and skilled interpretation of these before the diagnosis of ARVC is made.

Treatment for ARVC

There is currently no known cure for ARVC. Treatments are currently directed at prevention of sudden death and collapse that result from rapid ventricular arrhythmias, which render the heart incapable of maintaining circulation of blood to the brain and body, and reducing symptoms of palpitations.

Treatments for ARVC include medications to help stabilise the heart rhythm and prevent the heart from beating too vigorously, your doctor will advise you which medications are best for you. Implantable defibrillators (ICDs) may be implanted into people who are felt to be at high risk, these devices detect rapid erratic heart beats and give a 'shock' which allows the heart's own pacemaker to take over and control the heart rhythm again. Devices such as the implantable defibrillator are usually used in conjunction with medications.

Other treatments may include radio-frequency or cry-ablation (which removes the sources of electrical short circuits inside the heart). Electrical short circuits can give rise to rapid heart rhythms and so elimination of these areas by ablation can remove them and stop dangerous heart rhythms arising. Your doctor will advise you which treatments are recommended in your case.

Your doctor may also ask you to refrain from intense physical activity, heavy manual labour and competitive sports.

Additional information

If you would like more information about ARVC / ARVD, the following websites may be useful.

www.arvd.com

www.arvd.net

www.arvd.org

www.cidg.org

Participation in Cardiac inherited disease registry:

If you would like more information prior to deciding whether you would like to join the Cardiac inherited disease registry, please feel free to contact us.

If you have any queries or concerns regarding your rights as a participant in the Cardiac Inherited disease registry you can contact an independent Health and Disability Advocate. This is a free service provided under the Health & Disability Act; through the offices of the Health and Disability Commissioner.

Telephone (NZ wide): 0800 555 050

Free Fax (NZ wide): 0800 2787 7678 (0800 2 SUPPORT)

Email: advocacy@hdc.org.nz

**This N.Z Registry has received Ethical Approval from the N.Z. Multi-centre Ethics Committee:
AKX/02/00/107**