



What is arrhythmogenic cardiomyopathy?

Arrhythmogenic cardiomyopathy (AC), previously referred to as arrhythmogenic right ventricular cardiomyopathy (ARVC) or arrhythmogenic right ventricular dysplasia (ARVD), is a rare type of cardiomyopathy that typically does not appear until teenage years or adulthood.

AC causes part of the myocardium (muscular wall of the heart) to break down over time. The muscle tissue in the **ventricle** dies and is **replaced with fatty scar tissue**. As the scar tissue builds up in the walls of the ventricle, ventricular dysfunction, or a decreased ability of the heart to pump blood out to the body, may result.

The result of these structural changes

can cause the electrical signals that control the heartbeat to be disrupted, causing arrhythmia (abnormal heart-beat). This disruption in the heart's electrical signal can lead to an increased risk of sudden cardiac arrest.

How many children are affected?

AC is the least common type of cardiomyopathy in children. It has been estimated that AC occurs 1 in 2,500 people, though it may be underdiagnosed because it is difficult to detect in people with mild or no symptoms. AC is very rarely recognized in children under the age of 10 years old.

What is the prognosis?

The outcome of children with AC is variable. AC is a rare disease, which has not been well studied in the pediatric population. Some children will need advanced therapies, such as a ventricular assist device or heart transplantation, but overall, there is limited information on treating children with the disease.

This booklet was created to provide families and caregivers with a broad overview of cardiomyopathy and is for general information only. The material presented is not intended to be complete or serve as medical advice. The information should not be a substitute for consultation with a qualified health care professional who is more familiar with individual medical conditions and needs.

The Children's Cardiomyopathy Foundation (CCF) is dedicated to finding causes and cures for pediatric cardiomyopathy through the support of research, education, and increased awareness and advocacy.

Register with CCF for additional resources:
childrenscardiomyopathy.org



Understanding



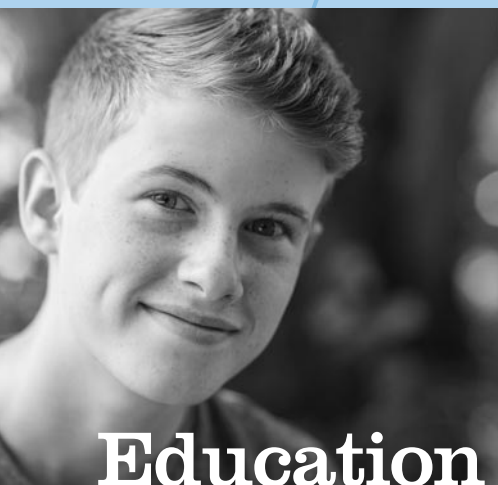
Children's Cardiomyopathy Foundation
24 West Railroad Ave., Suite 408, Tenafly, NJ 07670
Tel: 866.808.CURE (2873) • childrenscardiomyopathy.org
Email: info@childrenscardiomyopathy.org

Understanding Arrhythmogenic Cardiomyopathy



What causes AC?

The exact cause of AC is not always known, but it is believed to be an inherited disease. In 50 percent of cases, the disease runs in the family. Most familial cases of the disease are inherited in an autosomal dominant manner, in which one parent contributes a mutated gene and there is a 50% chance the child will inherit the condition. In rare instances, AC has an autosomal recessive pattern of inheritance, which means that both parents each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.



Education

AC can result from mutations in multiple genes, many of which are known as desmosomal genes. Desmosomal genes provide instructions for making components of cell structures called desmosomes, which attach heart muscle cells to each other. If desmosomal genes are mutated, their function is affected, causing cells of the myocardium to detach from one another and die. Over time, the damaged myocardium is replaced by fat and scar tissue. Gene mutations have been found in about 60% of AC patients, with the most common mutations in a desmosomal gene called PKP2.

AC also can occur secondary to Naxos syndrome and Carvajal syndrome, which are rare disorders that affect the skin, hair, and nails.

What are the common symptoms?

AC may not cause any symptoms in its early stages. Since it is a progressive condition, symptoms usually worsen over time. Symptoms are generally related to the electrical activity of the heart as well as how well it pumps. Since AC impacts the electrical signaling in the heart, children may experience **arrhythmia** (irregular heartbeat). This may put children at a higher risk of sudden cardiac arrest, especially during strenuous exercise.

The more common symptoms of AC include palpitations, a sensation of the heart skipping a beat, fluttering, or pounding in the chest, and presyncope with light-headedness or dizziness. In some cases, loss of consciousness or fainting (syncope)

may occur because arrhythmias can lead to reduced oxygen levels or blood flow to the brain.

If the myocardium becomes severely damaged in the later stages of AC, it can lead to heart failure. At this point, the symptoms are consistent with dilated cardiomyopathy. Symptoms that may develop include swollen legs, ankles, or stomach (edema), or there is shortness of breath and difficulty breathing (dyspnea)

How is AC diagnosed?

Generally, AC can be difficult to diagnose because changes in the heart can be subtle and the scar tissue can be hard to see. Additionally, since the heart chambers can become enlarged, it is sometimes misdiagnosed as dilated cardiomyopathy (DCM). AC is typically not diagnosed under the age of 10. Since symptoms of the disease are usually not present until later in life, it is more commonly diagnosed between 20 - 40 years of age.

Arrhythmias in the heart ventricles is a serious concern particularly in children and adolescents. They must be carefully evaluated and followed up especially when there is a family history. To access symptoms and make a diagnosis, a patient's medical history is first examined. It is important to look for other family members who may have the disease, as the condition can be inherited or genetic.

Patients may undergo a physician examination, **electrocardiogram** (ECG) to look at the electrical activity of the heart and check for arrhythmias, or an **echocardiogram** (ECHO) to examine the structure of the heart and how it is functioning. A magnetic resonance

imaging (MRI) scan may also be used to produce a high-quality image of the heart and blood flow.

Additional tests that may be conducted include an **exercise stress test** to see how well the heart handles exertion and **Holter monitoring** (when the ECG is recorded over a period of time) to monitor a patient during their daily activities. An electrophysiology test (EPS) involves having a long tube (catheter) placed into a blood vessel and fed to the heart. Electrical signals are sent to the heart from the catheter to make it beat at different rates. This is recorded and used to determine the presence of arrhythmias and subsequent treatment options.

An implantable **loop recorder** is another method that may be used to assess symptoms. This small device is implanted under the patient's skin and records the activity of the heart to identify arrhythmias. Loop recorders can be implanted for several years if needed.

What are the treatment options?

Cardiomyopathy is a variable disease without a cure. Therefore, treatment plans will vary with each patient and management of the disease is aimed at minimizing and controlling symptoms and reducing the risk of complications. Like other forms of cardiomyopathy, treatment for AC is focused on improving the pumping of the heart, controlling arrhythmias, and reducing the risk of cardiac arrest.

Common treatment options of AC include **medications** such as anti-arrhythmic drugs to reduce abnormal heart rhythms and, anticoagulants to reduce the risk of blood clots forming.



Treatment

If the disease is more advanced and there is heart failure, other medications may include Angiotensin Converting Enzyme Inhibitors, Angiotensin Receptor Blockers, and/or beta blockers which help to lower the heart rate. Other medications to treat arrhythmias may also be used. For patients with symptoms of heart failure caused by fluid retention, diuretics, which are water tablets that reduce the buildup of fluid in the body, can be prescribed.

Other interventions for patients with increased symptoms and at risk or sudden cardiac arrest may include an **implantable cardioverter defibrillator** (ICD) or in rare cases a catheter ablation. The ICD is the most effective safeguard against sudden cardiac death. An ICD may offer a survival benefit of up to 50% in patients with AC. An ICD is a device powered by a battery, placed under the patient's skin with thin wires that connect the ICD to the heart. The ICD keeps track of their heart rate, and if an abnormal heart rhythm is detected, the ICD will deliver an electric shock to restore the heart to a normal heartbeat.

In **catheter ablation**, the areas of the heart causing arrhythmias are located and cauterized (burned) to destroy the scar tissue. This is an invasive procedure and is performed in an electrophysiology laboratory. While this procedure may reduce the frequency of arrhythmic episodes, it does not cure the underlying problem with AC.

There are risks associated with ICD implantation as well as catheter ablation, so it is important to have a detailed discussion with a pediatric cardiologist to determine the most appropriate course of management. In patients in whom AC has progressed to heart failure, a heart transplantation may be recommended.

In addition to medication and devices, a healthy lifestyle is an important part of disease management, including eating a balanced diet, maintaining a healthy weight, minimizing alcohol, and not smoking. Exercise may need to be modified because AC can trigger dangerous arrhythmias and heart failure symptoms. Physical activity guidelines may need to be discussed with the medical team to ensure safety.