Antiarrhythmic Medications

If you continue to have symptoms of ACM even while taking beta-blockers, your cardiologist may recommend that you take antiarrhythmic medication as well. These medications reduce the likelihood of an arrhythmia. Some types of antiarrhythmic medications that are used in ACM are amiodarone (brand name Cordarone), sotalol (Betapace), flecainide (Tambocor), propafenone (Rhythmol), and mexilitene (Mexilitil). It's important to understand how to take them properly and discuss your concerns with your cardiologist before starting them.

Implantable Cardioverter Defibrillators (ICDs):

In some people with ACM, the heart may suddenly start beating too rapidly to properly pump blood to the body. During this fast rhythm, a strong electric shock to the heart is the only way to "reset" it so it can start beating in a normal rhythm again. The process of shocking the heart back into a normal rhythm is called defibrillation.

ICDs are recommended in ACM patients who still have an irregular heartbeat despite medical therapy, or who have a family history with relatives who died suddenly of ACM. They are also used as first-line treatment in people who experienced a cardiac arrest and had to be resuscitated.

Many people have difficulty adjusting to the idea of an object inside their body. For these reasons, it is important to address all of your concerns, including emotional concerns, with your doctor.

Radio Frequency Catheter Ablation

This treatment is usually recommended for ACM patients who do not respond well to antiarrhythmic medications, or patients with an ICD that is shocking too often. The abnormal heart rhythms in ACM occur when heart muscle is slowly replaced by scar tissue and fat, which conducts electrical signals differently than normal heart muscle. Destroying tiny areas of tissue that cause this dangerous electrical signal can prevent abnormal heart rhythms.

Treatment for Heart Failure

Heart failure is when the heart becomes weak and is no longer able to pump blood effectively. There are many causes of heart failure, but in patients (rarely children) with ACM, it is due to the replacement of muscle with fat and scar tissue. Symptoms of heart failure include leg swelling, cough, chest pain, trouble exercising, and shortness of breath. If you are experiencing these symptoms, your cardiologist may prescribe medications to help strengthen the heart or to reduce its workload.

Treatment of Asymptomatic Patients and Family Members

Asymptomatic patients and family members do not require specific antiarrhythmic or other cardiac treatment. However, they should be followed by regular non-invasive cardiac investigations for early recognition of disease.

What lifestyle changes can I make to help my condition?

In addition to your medication, surgery, and/or ICD, there are some lifestyle changes you can make to reduce your chance of having an episode of arrhythmia. The same recommendations are not necessarily appropriate for every patient. Ask your doctor which lifestyle modifications they recommend for you.

Participation in Sports

Because dangerous arrhythmias are more likely to occur when the heart is beating rapidly, strenuous and competitive sports, or endurance or vigorous exercise are not recommended for people with ACM. Ask your cardiologist which activities are appropriate for the severity of your condition. Lower energy sports such as walking, golf, bowling, and curling may be good ways for people with ACM to stay active without increasing their risk. It's very important to discuss your child's physical limitations with their school.

Medications to Avoid

Some medications cause the heart to beat more quickly as well, and these should be avoided. These medications include stimulants like Sudafed (pseudoephedrine), which is a non-drowsy allergy medicine and can be purchased without a prescription. Always ask your doctor or pharmacist before taking any medication.

What are some parenting challenges when raising a child with ACM?

Raising a child can be difficult, and raising a child with ACM presents a whole new set of challenges. Many parents find that learning more about the condition helps them to feel more in control. Some people find it helpful to speak with other parents, patients, or siblings, who have had similar experiences. The SADS Foundation website (StopSADS.org) is a great place to read people's stories, connect with others, and share your own experiences.

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SADS FOUNDATION

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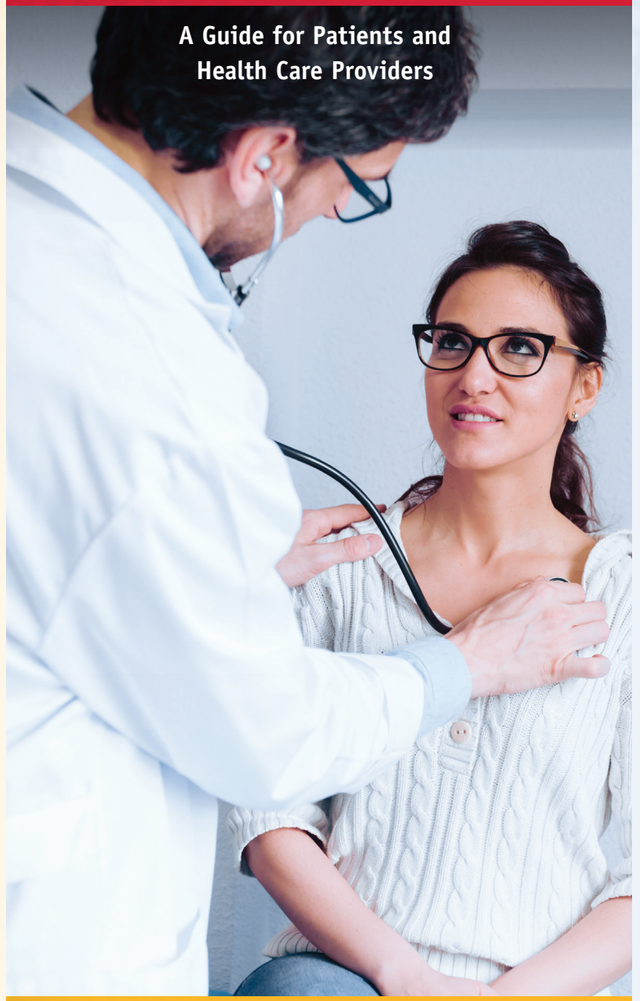
Arrhythmogenic Cardiomyopathy (ACM) or Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

A Guide for Patients and Health Care Providers



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Why do I need to know about Arrhythmogenic Cardiomyopathy?

Arrhythmogenic cardiomyopathy, or ACM (formerly known as arrhythmogenic right ventricular cardiomyopathy, ARVC, or arrhythmogenic right ventricular dysplasia, ARVD), is a genetic disease of the heart muscle. Although most people with ACM do not show any symptoms, the condition causes 15%-25% of heart-related deaths in people under 35 years of age. The exact prevalence of ACM is unknown but is estimated to be between 1 in 5,000 to as high as 1 in 1,000 people.

ACM can be diagnosed early and sudden death prevented. Still, this condition is often undetected before death and may not be recognized as the cause of death. Family members of individuals with unexplained death should be tested for ACM and other genetic heart conditions. If an inherited condition like ACM was responsible for the unexplained death, then 50% of family members may also be affected. ACM is a treatable disorder and, with correct diagnosis and common treatments, most deaths are preventable.

Physicians need to know:

- When to consider ACM as a possible diagnosis.
- When to refer patients for diagnosis & treatment.
- About genetic testing for ACM and other SADS conditions.
- How to develop a family pedigree and screen family members for ACM.

What is ACM?

Arrhythmogenic means "causing the heart to beat irregularly," and the word cardiomyopathy is translated as "heart-muscle disease." Physicians and scientists have discovered that ACM is caused by a defect in the tiny proteins that hold the heart muscle cells together. These complex proteins or "desmosomes" can be disrupted in ACM, leading to areas of scar and fat deposits. This scarring can lead to abnormal heart rhythms which can cause patients to become symptomatic and sudden cardiac arrest or death.

Ventricular tachycardia is an abnormal rhythm that causes the heart to beat too fast to pump blood effectively out of the heart, and the person may lose consciousness within seconds. This sudden loss of consciousness is called syncope and may look like a simple faint or like a seizure. ACM patients who collapse for this reason may recover on their own or they may require CPR (cardiopulmonary resuscitation) or defibrillation (an electric shock to the heart delivered by an AED or Automated External Defibrillator) to start the heart beating normally again.

The symptoms of the disease mostly occur in adults in their 30's and 40's. The first signs of ACM may be subtle and are easily missed. This is why family members of people with ACM must undergo extensive testing to check if they are affected by the condition.

ACM is thought to progress in several stages. The first stage is a "quiescent phase" where the patient has no symptoms, but still may have a risk of sudden death, as ECG abnormalities do occur particularly during heavy exercise. Patients experience symptoms in the second, or "overt" phase. In this phase, the ventricles may have episodes of arrhythmia (irregular beating), which are perceived as dizziness, heart palpitations, and fainting. Other more non-specific symptoms like fatigue, confusion, and abdominal pain may be present as well.

In the later phase, symptoms of heart failure, such as swelling of the legs, fluid in the lungs, and shortness of breath, may develop. Because ACM is progressive, and can show up at any time, repeat testing of people at risk is necessary. Early diagnosis is important because in some people the first symptom of ACM is sudden cardiac arrest.

What are the symptoms of ACM?

Patients may present with any one or more of the following symptoms:

- Palpitations (abnormality of heartbeat that causes a conscious awareness of its beating)
- Syncope (fainting or seizure-like activity)
- Light-headedness
- Chest pain
- Cardiac arrest

Who should be assessed for ACM?

- When there is a **family history** of relatives known to be affected by ACM.
- People with known relatives who have a gene variant (mutation) consistent with ACM.
- People who have **fainted or had seizures during exercise or emotional excitement.**
- People who had relatives who died suddenly at a young age (under the age of 40).
- People who present with a **ventricular arrhythmia not caused by heart blockages** or myocardial infarction.

How is the diagnosis made?

ACM is difficult to diagnose, which can lead to frustration on the part of patients, families, and even their doctors. Even after many tests, your doctor may not be able to say for sure whether or not you or your child has ACM. Cardiologists use a list of criteria, or scoring system, to make a clinical diagnosis of ACM. An important criterion in the scoring system is family history, so it is important to know if any relatives died suddenly at a young age, or had unexplained heart arrhythmias.

Testing for ACM involves a combination of tests. The number and types of tests you will have to undergo depend on how definitive the results of earlier tests are. People at high risk will often be assessed every year or two to determine if signs of ACM have developed. Many of these tests are not available at all hospitals, so you may have to travel to a larger medical facility to have them performed.

Tests Used to Evaluate for ACM: Electrocardiogram (ECG), SAECG, Heart Rhythm Monitoring and Stress Testing

An ECG machine measures the electric pattern of the heart through electrodes placed on the skin. The detailed ECGs used for the diagnosis of ACM involve ten or more electrodes on the chest, arms, and legs.

Some people with ACM have abnormal ECGs when they are at rest, and sometimes the irregularity is only apparent when the person is feeling palpitations or lightheadedness. Since these episodes don't always happen at the doctor's office, you may be given a heart rhythm monitoring device called a Holter monitor or Event looping monitor to take home. These small portable ECG machines can be worn at home for 24 or 48 hours or up to 30 days to detect arrhythmias that occur during daily activities.

An exercise stress test will be part of diagnostic testing for ACM. In this test, a person is hooked up to an ECG machine while they run on a treadmill or ride an exercise bicycle. This test can be useful in diagnosis because the heart is more likely to have an unusual rhythm when it is beating quickly.

Echocardiogram

An echocardiogram is an ultrasound test that determines the size and shape of the heart. With ACM, the ventricles, or bottom chambers of the heart may be enlarged and this can be picked up during the echocardiogram. Abnormalities from the echocardiogram are not enough to diagnose ACM, but are part of the diagnostic puzzle of the criteria and scoring system for ACM.

Cardiac MRI

Cardiac MRI (magnetic resonance imaging) is a good way to visualize the changes in the heart that can occur with ACM and is another tool used in the diagnostic puzzle.

Like echocardiography, an MRI is a non-invasive test that does not involve radiation. Abnormalities in the size, shape, movement or tissue composition of the heart can be detected if present. Cardiac MRIs are not performed commonly at most hospitals and can be misread if not done at an institution with expertise in both performing and interpreting MRIs for ACM. The SADS Foundation can help you find a qualified health care facility to perform this important diagnostic tool.

Electrophysiological Testing

Electrophysiology testing is an invasive test that is sometimes performed both to diagnose and treat ACM. This test is typically performed under sedation or with anesthesia. The origin of the irregular beats can be identified, aiding the doctor in future risk assessment, placement of an ICD, or the need for catheter ablation.

What about genetic testing?

ACM is caused by a genetic variant, or change in the genetic instructions that is different from the reference code. Therefore genetic testing is part of the diagnostic criteria and can help in making a diagnosis.

Most ACM patients inherit the condition from an "autosomal dominant" pattern, meaning that only one parent had the gene. Members of the same family can share the same genetic mutation, but one case can be severe ACM and the other may have no symptoms at all.

Gene variants can be identified in some, but not all cases of ACM, so a negative ACM gene mutation does not rule out the condition.

The SADS Foundation can help identify a certified genetic counselor to walk you through this process. Visit StopSADS.org for more information.

How is ACM inherited, and who in a known or suspected family should be tested?

ther family members of ACM patients should be genetically tested and screened for ACM. When a patient is clinically identified as having ACM and the patient has tested positive for a specific ACM gene, care should be taken, with the help of a genetic counselor, to determine who else in the family should be genetically tested.

Some ACM patients do NOT have a positive gene mutation, but first degree family members (parents and siblings) should still be seen by a cardiologist to rule out ACM. Knowledgeable genetic counselors and cardiologists can help families interpret the meaning of genetic tests and decide what precautionary measures should be taken for extended family members.

What is the treatment and who should be treated?

There are many different treatments available, and which one is appropriate in a given situation depends a great deal on the particular patient.

Beta-Blockers

In ACM, medication cannot cure the underlying disease, but may relieve symptoms like palpitations and reduce the chance of a fatal episode. Standard treatment for ACM is a class of medications called beta-blockers. Beta-blockers, with names like atenolol, metoprolol, and nadolol, are frequently recommended. Some of the side effects of beta-blockers are slow heart rate, fatigue, dizziness, drowsiness, cold feet, sleep problems, and weight gain. Sometimes it takes a while for the body to adjust to beta-blockers, and your doctor may have to adjust your dose or change medications to deal with side-effects. Also, as a child grows, his or her beta-blocker dose will usually have to be increased to adjust to their increase in weight and size.

Once you are taking beta-blockers, **you must take the prescribed pills every day, since stopping the medication suddenly can put you at an even higher risk for an arrhythmia**. There are different types of beta-blockers, so talk to your cardiologist if you find the side effects of the medication unacceptable.