Patient’s Guide to Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
Past to Present

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Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) is an inherited progressive disease of the heart muscle that can cause dangerous heart rhythms (arrhythmias). Significant advances over the past 10 years have changed how we diagnose and manage patients with ARVD/C and at-risk family members. In this guide, we review key information that every family affected by ARVD/C should know (Table 1).

The majority of patients with ARVD/C have abnormalities of the right ventricle. However, abnormalities of the left ventricle are more common than once thought. Recently, the main areas of the heart involved in ARVD/C, referred to as the triangle of dysplasia, have been redefined as noted in Figure 1.1 ARVD/C, if severe, may impair the ability of the heart to pump blood as needed. ARVD/C is a genetic disease and often runs in families, making evaluation of first-degree relatives essential.

What Is the Cardiac Desmosome?
It has been shown that ARVD/C is often a disease of the cardiac desmosome. Desmosomes are proteins that connect heart muscle cells and hold them together (Figure 2). Changes (mutations) in the genes (Plakophilin-2 [PKP2], Desmoglein-2 [DSG2], Desmocollin-2 [DSC2], Plakoglobin [JUP], Desmoplakin [DSP]) that make desmosome proteins can result in weakened desmosomes.2,3 When this happens, heart muscle cells become disconnected and die, particularly when the heart is stretched during vigorous exercise. Damaged heart muscle is ultimately replaced with fat and scar tissue.

Should I Have Genetic Testing Done?
Genetic testing of the desmosomal genes is now available through several clinical laboratories. Genetic tests including even more genes associated with ARVD/C, as well as other inherited heart diseases, are available and may be recommended by your healthcare provider.7 It is strongly recommended that patients interested in pursuing genetic testing discuss the risks, benefits, and limitations of genetic testing with a genetic counselor (Table 2).

Genetic testing can be helpful in confirming the diagnosis of ARVD/C in someone with signs and symptoms. Once a genetic change has been identified in an individual with ARVD/C, carrier testing can be offered to family members. This can help determine who is at increased risk of developing ARVD/C in the family. A negative genetic test result in someone with signs and symptoms of ARVD/C does not rule out the diagnosis because not all genes involved in ARVD/C have been identified. For family members, it is important to understand that a genetic change indicates the potential to develop ARVD/C and not necessarily a diagnosis of ARVD/C. Not everyone...
with a genetic mutation will develop signs and symptoms of ARVD/C.

**Updated Diagnostic Criteria: 2010 to Present**

The clinical diagnosis of ARVD/C is guided by the Task Force Criteria, which were revised in 2010. These criteria are more sensitive in identifying mild and early cases and now incorporate genetic testing. They look for abnormal changes in the structure and function of the right ventricle, as well as arrhythmias, family history, and genetic testing results. To apply the Task Force Criteria, patients should be evaluated with several noninvasive cardiac tests (Table 3). The diagnosis of ARVD/C is never based on a single test. On the basis of the results of multiple tests, additional testing may be recommended, including genetic testing.

**Can ARVD/C Be Treated?**

The primary goals of current ARVD/C treatment strategies are to protect patients from life-threatening arrhythmias, to reduce arrhythmias and stress on the heart, and to prevent progression of the disease (Table 4).

Most patients with ARVD/C undergo implantation of an implantable cardioverter-defibrillator (ICD) to treat dangerous rhythms. ICDs are very successful in preventing sudden cardiac death in ARVD/C. For a detailed explanation of an ICD, please see the Cardiology Patient Page article by Reiffel and Dizon.

Medication, including β-blockers, antiarrhythmics (sotalol, amiodarone, etc.), and angiotensin-converting enzyme inhibitors are regularly used to reduce arrhythmias and stress on the heart. Medication alone may not be enough to prevent arrhythmias. Clinical trials are being developed to assess new therapies such as the use of diuretics and preload-reducing therapies that may be able to prevent the progression of ARVD/C.

Advances in catheter ablation (caterization of areas of the heart to reduce arrhythmias) and our understanding of where arrhythmias are located within the heart have resulted in improved techniques and treatment results. An endocardial approach treats arrhythmias coming from the inside of the heart, and an epicardial approach treats arrhythmias on the outside of the heart. Whereas catheter ablation is not considered curative, it does help reduce possible life-threatening arrhythmias. Because the disease usually begins from the outer layers of the heart, the epicardial approach has been shown to be more effective but must be done at specialized, experienced centers because the procedure has significant risks.

In rare cases, patients with ARVD/C develop severe heart failure or incessant arrhythmias, and a cardiac transplantation may be necessary. Although there have been significant improvements in the diagnosis of ARVD/C and how to identify at-risk family members, there remains a lack of information about how to treat ARVD/C more effectively. Research is ongoing using a special type of stem cell to model ARVD/C in the laboratory, allowing researchers to better understand the disease process and subsequently develop new therapies that could potentially halt progression.

**I Am at Increased Risk of Developing ARVD/C; What Do I Do?**

First-degree family members and those who carry a genetic change but have not been diagnosed with ARVD/C should undergo cardiac screening every 2 to 3 years starting between 10 and 12 years of age. The frequency of repeat screening is based on age, family history, athletic history, and cardiac test results and should be discussed with your physician. A recent study from our group has shown that a person’s electrocardiogram and Holter monitor results can help predict their risk of arrhythmias and need for an ICD. Family members with abnormal electrocardiograms and...
high premature ventricular contraction count on Holter are at highest risk of developing arrhythmias.11

**I Have ARVD/C; Can I Exercise?**

Participation in competitive endurance-type athletics has been associated with increased risk of developing ARVD/C, having arrhythmias, and having disease progression. Recently, it has been shown in patients that both participation in vigorous endurance athletics and greater duration of exercise per year are associated with an increased likelihood of being diagnosed with ARVD/C and developing ventricular arrhythmias and heart failure.12 For this reason, it is recommended that patients with ARVD/C avoid participation in most sports and avoid strenuous exercise.

**I Have ARVD/C; Can I Have Children?**

ARVD/C can run in families, being passed from one generation to the next. Patients diagnosed with ARVD/C are faced with the psychosocial complexities of living with and having a family with an inherited condition. Meeting with a genetic counselor is helpful in discussing options for family planning. ARVD/C is not a contraindication for having children per se because the phenotype is influenced by environmental factors that can be modified to a certain extent. Furthermore, because the diagnosis and treatment of ARVD/C have improved, many women with ARVD/C have had healthy, uneventful, full-term pregnancies.13 It is important to make sure that arrhythmias are well-controlled before pregnancy and plans for a safe delivery are in place. Current recommendations for pregnancy management include an electrocardiogram, an echocardiogram, and 24-hour Holter monitoring as soon as the pregnancy is detected and again at 3 months postpartum, as well as a repeat 24-hour Holter monitor at 7 months of pregnancy. In addition, the potential harmful effects of certain cardiac medications during pregnancy should be considered before starting a family.

**Living With ARVD/C**

As with any chronic illness, there are challenges to adjusting to life with a new diagnosis. ARVD/C is no exception. ARVD/C presents at a much younger age compared with most heart diseases. It also is associated with a high risk of potentially life-threatening arrhythmias. As a result, many patients with ARVD/C undergo placement of an ICD. It is recognized that young patients with ARVD/C have a higher rate of device-related anxiety and body-image concerns.13,14 Referral to mental health services should be widely offered to help with adjustment and to improve quality of life.

Because ARVD/C is often diagnosed in high-level competitive athletes, it is challenging to move away from a lifestyle that is now considered harmful. A diagnosis of ARVD/C poses many challenges not only to the individual living with the diagnosis but also to the family members who may be at increased risk of developing ARVD/C. Managing the emotional aspect of ARVD/C is just as important as identifying a medical team experienced with the diagnosis and management of ARVD/C. Our long-ranging aim is the development of medical therapy to cure the disease or at least to modify its overall course in a more favorable way. Once an appropriate management plan is in place and patients become empowered through knowledge and increased confidence, most are able to live a long and fulfilled life (Table 5).

**Additional Resources**


**Sources of Funding**

We acknowledge funding from the Dr Francis P. Chiaramonte Private Foundation, St. Jude Medical Inc. and Medtronic Inc. The Johns Hopkins ARVD/C Program is supported by the Leyla Erkan Family Fund for ARVD Research, the Dr Satish, Rapal, and Robin Shah ARVD Fund, the Dr Satish, Rapal, and Robin Shah ARVD Fund.
Disclosures
Dr Sears serves as a consultant to Medtronic and has received research grants from Medtronic in past 2 years. These funds are directed to East Carolina University. Dr Sears also has received speaker honoraria from Medtronic, Boston Scientific, St. Jude Medical, and Biotronik. The other authors report no conflicts.

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*Circulation*. 2014;130:e89-e92
doi: 10.1161/CIRCULATIONAHA.113.004845
*Circulation* is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 0009-7322. Online ISSN: 1524-4539

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