Johns Hopkins Arrhythmogenic Right Ventricular Dysplasia Program

Volume 1, Issue1 Winter 2011

Introduction

We are excited to present our first newsletter to be published semi-annually. For those of you who are new to the program, The Johns Hopkins Arrhythmogenic Right Ventricular Dysplasia Program was established in 1999 through the combined efforts of several families affected by this condition and a team of cardiologists from Johns Hopkins. The mission of the Johns Hopkins ARVD Program is three-fold:

1) **To provide education for patients and physicians about ARVD**

2) **To facilitate the evaluation and management of patients with known or suspected ARVD.**

3) **To provide new knowledge of ARVD**

The program is led by Dr. Hugh Calkins, MD, Professor of Medicine and Pediatrics and Director of the Arrhythmia Service and the Clinical Electrophysiology Laboratory. The ARVD team consists of three genetic counselors (Cynthia James, ScM. PhD; Brittney Murray, MS; and Crystal Tichnell, MGC). Dr. Harikrishna Tandri, MD is actively involved with the ARVD program seeing patients and performing electrophysiology procedures. Dr. Dan Judge, MD heads the genetic research and sees patients with heart failure symptoms.

Education

The gateway to knowledge of ARVD is through the program's website, www.ARVD.com. The site is currently managed by the genetic counselors and is updated on a regular basis with new articles and questions relating to ARVD. The website offers an online registration form where individuals with ARVD can enter their information for the Johns Hopkins Registry and to see if they are candidates for other ARVD-related research projects being conducted at Johns Hopkins. Individuals can also request a free brochure on ARVD and email questions to a genetic counselor.

The Johns Hopkins ARVD Program hosts an annual conference for those affected with ARVD, their family members, faculty, colleagues, and other interested parties. Exciting new research is presented and attendees have the opportunity to interact with the leading ARVD clinicians and researchers. This also provides an important opportunity for families affected by ARVD in interact with each other. We also provide the opportunity for those with ARVD to participate in research studies during this time.

Patient Care

The ARVD Program staff is available for formal consultations and second opinions regarding a diagnosis or possible diagnosis of ARVD, to discuss management of ARVD, and to undergo genetic counseling and testing. Patients are seen in consultation with Dr. Hugh Calkins or Dr. Hari Tandri and one of the genetic counselors to discuss test results, family history and to provide guidance regarding further management. Appointments with our heart failure specialists, Drs. Stuart Russell and Daniel Judge, are also available. In selected cases we also offer ablation as a treatment for difficult to manage ventricular tachycardia. We can also arrange for appropriate screening for family members.

Our program has worked with individuals from all over the US and from throughout the world to either confirm a possible diagnosis of ARVD or for advice regarding treatment.

To schedule an appointment, contact Crystal at 410-502-7161 or ctichnell@jhmi.edu.

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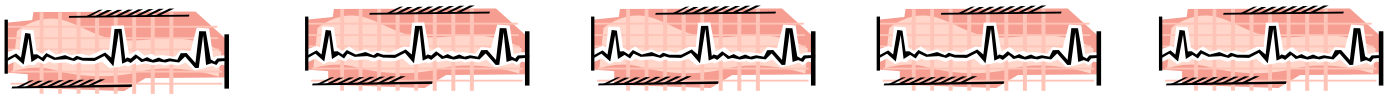
Research

The heart of our program is the Johns Hopkins ARVD Registry. We use the Registry as the basis for most of our studies on diagnosis, treatment, genetics, and progression. The major goal of our genetic research is to locate new genes associated with ARVD and to study the relationship between different genetic causes of ARVD and the clinical features of the disease. Participation in the registry involves submitting medical records and/or autopsy reports, questionnaires, submitting a blood sample for genetic research, and annual telephone follow-up.

In addition, we have received funding from St. Jude, Medtronic, and Boston Scientific Corporations to study arrhythmias in people with definite ARVD, people with probable ARVD, and their family members.

Our program has also been working hard over the last few months on abstract submissions to the Heart Rhythm Scientific Session in May. These abstracts include our analysis of the efficacy of endocardial catheter ablation procedures in patients with ventricular tachycardia and ARVD, the incidence and predictors of appropriate ICD therapy in patients with ARVD undergoing ICD implantation for primary prevention, a re-evaluation of signal averaged electrocardiography in ARVD, the penetrance of desmosomal mutations in familial ARVD, and a review of patients requiring heart transplant in the Johns Hopkins Registry aiming to better understand the indications and risk factors for cardiac transplant. Results of these studies will be discussed as the research is completed and published.



Research Opportunities at Johns Hopkins

Clinical and Genetic Investigations of Right Ventricular Dysplasia (Registry)

Who: Children and adults with ARVD

What: Collection of pertinent past medical records and continued collection for 5 years. A blood sample for DNA for genetic mapping of ARVD genes

How to Join: Contact Crystal Tichnell, MGC at 410.502.7161 or ctichnell@jhmi.edu. She will need to send you a consent form, then review the submitted records and make arrangements for obtaining and shipping the blood sample.

Predictors of ICD Firing in ARVD Patients

Who: People with a definite diagnosis of ARVD and an implantable cardioverter defibrillator

What: This study is trying to learn more about what causes arrhythmias that need treatment with a shock from your ICD. You will be asked to answer some background questions about your diet, exercise and medications. If your ICD delivers a shock, you will be asked to answer additional, more detailed questions about your activities in the days before the shock. In addition, we will request copies of the ICD interrogations in order to learn more about the details of the arrhythmia.

How to Join: Contact the ARVD Clinic Office at 410-502-7161

Have you had an epicardial ablation?

We are looking for people with ARVD who have had an epicardial ablation to join our Registry. Help us discover how this new technique affects the course of ARVD!

Please contact Crystal (ctichne1@jhmi.edu; 410-502-7161) or Cindy (cjames7@jhmi.edu; 443-287-5985) if you are interested.

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Genetic Testing

Genetic Testing for ARVD is now offered by several clinical laboratories and includes 5-7 genes associated with ARVD, including PKP2, DSG2, DSC2, DSP, TMEM43, JUP, and RYR2. About 50% of patients who have ARVD will have an important change in one or more of these genes. Therefore, current genetic testing for ARVD is not definitive. Since we do not yet know all of the genes associated with ARVD, an individual who carries the diagnosis of ARVD may still test negative. A negative result does not exclude the diagnosis of ARVD. A positive result is certainly helpful in confirming the diagnosis as well as helpful in determining who in the family is at increased risk of developing ARVD. In some cases, the significance of a result is unknown and may require further investigation. We strongly recommend speaking with a genetic counselor prior to genetic testing so that you understand the risks, benefits, and limitations of ARVD genetic testing.

ARVD Genetic Testing Labs in the United States

- **GeneDx**—7 gene panel (www.genedx.com)
- **Harvard Laboratory for Molecular Medicine**—5 genes (www.pcpgm.partners.org/lmm)
- **PGxHealth**—5 gene panel (www.familion.com)
- **Correlagen**—5 gene panel (www.correlagen.com)

ARVD Genetic Testing Labs in Canada

- **The Hospital for Sick Children**—
- 4 gene panel (www.sickkids.ca)

Featured Publication

Marcus F., et al. **Diagnosis of ARVC/D: Proposed Modification of the Task Force Criteria**. *Circulation*. 2010; 1532-1541.
http://circ.ahajournals.org/cgi/content/full/121/13/1533

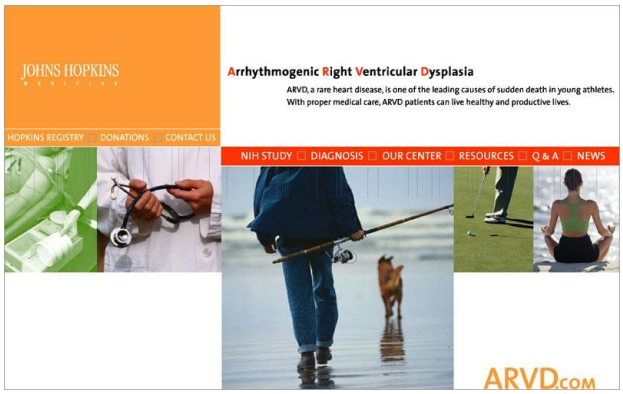
Data from 108 individuals with newly diagnosed ARVD, who were enrolled in the NIH-sponsored Multidisciplinary Study of ARVD were analyzed and compared with non-ARVD individuals. These individuals underwent ECGs, signal averaged ECG, Holter monitor, echocardiograms, MRIs, as well as some invasive tests. These results were carefully analyzed to determine criteria that were both specific and sensitive in order to improve the diagnosis and management of ARVD. The proposed modifications now take into account the presence of genetic mutations. Specific quantitative data must now be met in characterizing the size of the right ventricle. An enlarged right ventricle is no longer sufficient toward criteria. Signal averaged ECG criteria, as well as arrhythmia criteria have also been modified. Our program was involved in the modification of these criteria and we are in the process of reclassifying those enrolled in our registry.

ARVD Patient and Family Seminar

The next annual ARVD seminar will be held on Saturday, May 14th, 2011. Guest speakers include Dr. Jeffrey Saffitz, MD, a pathologist at the Beth Israel Deaconess Medical Center in Boston, MA and Dr. Samuel Sears, PhD, a professor of health psychology at East Carolina University in Greenville, SC. Our own Johns Hopkins speakers include: Hugh Calkins, MD; Stuart Russell, MD; Hari Tandri, MD; Cindy James, ScM, PhD; Brittney Murray, MS; and Crystal Tichnell, MGC. Once again, we will host a "Smoothie Social" on Friday, May 13th, 7-9pm. Details regarding hotel and travel are posted at www.ARVD.com. To register for the seminar, please do so online: http://www.arvd.com/conferences/family_seminar_2011.html

The morning part of the seminar will consist of several lectures and will conclude with lunch. During the afternoon session, we will hold a large question & answer session, along with opportunities to participate in research. We hope to see you in May!

ARVD Program Staff



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Dmitri Gagarin, MD – Researcher

Cynthia James, ScM, PhD—Genetic Counselor

Brittney Murray, MS—Genetic Counselor

Crystal Tichnell, MGC—Genetic Counselor

Looking for a support group?
Visit: www.arvd-arvd-info.com

How You Can Help

None of the research by the ARVD Program would be possible without the active participation of families affected by ARVD. To join our research, the first step is to enroll in the ARVD Registry and send us copies of your cardiac tests. We will then invite you to be a part of other research efforts that are appropriate for you.

We also rely on the generosity of families to financially support this program. Although we have applied for many grants from the NIH and other public funding organizations, we have had only limited success. In part, this reflects the fact that ARVD is a rare disease and not considered a major health hazard. It is critical for patients or families affected by ARVD to lobby for increased funding for research on this important condition.

If you are interested in making a financial contribution to the ARVD Program, please contact Shannon Curley, Director of Development for the Johns Hopkins Heart Institute at scurley3@jhmi.edu or 410-516-6607. You can also make an Online Gift through our encrypted, secure server at http://www.arvd.com/donations.html
If you are hosting an ARVD Fundraiser and would like to include information regarding your event in our newsletter, please email Crystal at ctichnell@jhmi.edu.

THANK YOU FOR YOUR SUPPORT!!!

Healing Hearts
Annual Bull & Oyster Roast
Friday, February 11, 2011 – 7pm-midnight
American Legion, 109 Dewey Lowman
1610 Sulphur Spring Road
Baltimore, Maryland 21227

Tickets cost $40 per person. Proceeds to benefit ARVD Research at Johns Hopkins Heart Institute. There will be raffles including a $500 grand prize, a silent auction and more! For more info, reserve a table or purchase tickets, please email HealingHeartsMD@yahoo.com
Visit www.HealingHeartsMD.com for info regarding this organization and events.

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