

JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

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* ** ***!!!HAPPY HOLIDAYS!!!*** ** *

We hope you and your families have had a wonderful Holiday Season and are looking forward to the New Year! This year has been no different. The ARVD/C Program has been hard at work with collaborators around the world. Thank you again for your dedication to the work of our program. Our work would not be possible without your continued enthusiasm and willingness to participate in our many research endeavors. As always, we are available for consultations and evaluations in order to determine the best management strategies for you. Please do not hesitate to contact our program with any questions or concerns. We look forward to seeing many of you at our upcoming Patient & Family Seminar in May 2014. Details are enclosed. Best wishes to each of you for a Healthy 2014!

~ The ARVD/C Program

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Mark your Calendars! 5/3/2014

It's that time of year again! Registration for the Annual ARVD/C Patient and Family Seminar is here! Lots of exciting information...meet the ARVD/C experts...meet other patients and families just like you...participate in research!

****New this year**** – *The ABC's of ARVD/C* – to be presented as an early morning session for new patients and new seminar attendees as a basic intro to understanding ARVD/C.

Meet Dr. Abhishek Sawant!

Dr. Sawant joined the ARVD/C Program in July 2013 as a post-doctoral fellow for the next two years. He completed his medical education in 2003 at one of the top 10 medical colleges in India. Subsequently he completed his Masters in Public Health from University at Buffalo School of Public Health with summa laude. He completed his Internal Medicine Residency at the University at Buffalo Medical and Dental Consortium Program and is currently Board Certified in Internal Medicine. Prior to joining the ARVD/C program, Dr. Sawant was an Internal Medicine Clinical Instructor at UCSF Fresno Medical Program for 5 years. His research interests include inherited cardiomyopathies and heart failure. He has also been studying the impact of exercise on ARVD/C patients and may have spoken to several of you regarding your exercise history. Welcome Dr. Sawant!



15th Annual ARVD/C Patient and Family Seminar

Presented by
The Johns Hopkins ARVD/C Program

You and your family members are invited to join us for our annual ARVD/C Seminar! You won't want to miss this unique opportunity to meet other families affected by ARVD/C and to learn the latest advances in the field. We are thrilled to have a number of guest speakers this year: Professor Cristina Basso, MD, PhD from Padova, Italy; Dr. Jeffrey Saffitz from Beth Israel Deaconess Medical Center in Boston, MA; and our own Director of Cardiology, Dr. Gordon Tomaselli. In addition, Dr. Dan Judge, Director for Inherited Heart Disease at Johns Hopkins will present an update on the Genetics of ARVD/C. Dr. Anneline te Riele, visiting PhD student from the Netherlands will also present her exciting research. After lunch, there will be an opportunity to ask questions in a "Question and Answer Session with the Experts", as well as an "Under 30" Discussion Group and opportunities to participate in research. Make plans to come into Baltimore early so you can attend a reception at the Hilton Garden Inn on Friday evening. Once again, we have an exciting and packed agenda so we look forward to seeing many of you in May! Specific details are enclosed. Tentative Agenda will be posted on our website under News and Events shortly. Please register early!

WHEN: Saturday, May 3rd, 2014 8:00am-5:00pm

WHO: Patients and Families affected by ARVD/C, Healthcare Professionals

WHERE: Chevy Chase Conference Center Auditorium, main level of Sheikh Zayed Tower at 1800 Orleans Street, Baltimore, Maryland 21287

COST: FREE

REGISTRATION: All participants must register! Please indicate on your registration form the names of other family members that are planning to attend. Register online by April 15th.

<https://www.surveymonkey.com/s/2014ARVCSeminar>

HOTEL ACCOMODATIONS – RESERVE EARLY!!!

Hotel rooms are available (limited) at the Hilton Garden Inn, Baltimore Inner Harbor (625 South President Street, Baltimore, Maryland 21202) at a special rate of \$139/night plus tax (single/double) until April 1st, 2014. Call 1-877-STAY-HGI or 1-877-782-9444 and mention the "ARVD Group" to receive the special rate. Check-in 3pm / Check-out 12pm. Hotel front desk phone is 410-234-0065. Self-Parking is available at a rate of \$23 and valet \$32 per day. www.baltimoreinnerharbor.hgi.com

TRAVEL TIPS

The Baltimore/Washington International (BWI) Thurgood Marshall Airport is the closet international airport to Johns Hopkins (www.bwiairport.com). It is approximately 30 minutes from the seminar location.

A bus will be available to transport seminar participants to the seminar on Saturday, departing only from the Hilton Garden Inn at 7:45am. The bus will leave the seminar to return to the Holiday Inn at 5:00pm.

Taxi Services – For Taxi services call Sun Cab at 410-235-0300 or Yellow Cab at 410-752-1096.

SPECIAL EVENT

Join us for a Meet 'n Greet Reception, 6:30-8:30pm, on Friday, May 2nd, 2014 in the Great American Grill at the Hilton Garden Inn. H'ors d'oeuvres will be served. Please register for this event when you register for the seminar or contact Crystal.

CLINIC CONSULTATIONS

Dr. Hugh Calkins and the genetic counselors will be available on Friday, May 2nd for consultations. Diagnostic tests can also be arranged if necessary. These appointments will be billed to your insurance. Please contact Crystal ASAP to schedule an appointment.

Abstracts and Presentations

The American Heart Association's Scientific Sessions is the leading annual convention for scientists and healthcare professionals devoted to the science of cardiovascular diseases and stroke, and the caring of patients suffering from these diseases. Its mission is to build healthier lives, free of cardiovascular diseases and stroke. The American Heart Association Scientific Sessions were held in Dallas, Texas in November 2013. Three abstracts were presented at this year's conference and two are summarized below.

MALIGNANT ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY WITH A NORMAL 12-LEAD ELECTROCARDIOGRAM: A RARE BUT UNDER RECOGNIZED CLINICAL ENTITY

Anneline S.J.M. te Riele, Cynthia A. James, Aditya Bhonsale, Judith A. Groeneweg, Christian F. Camm, Brittney Murray, Crystal Tichnell, Jeroen F. van der Heijden, Dennis Dooijes, Daniel Judge, Richard N.W. Hauer, Harikrishna Tandri, and Hugh Calkins

The goal of this study was to estimate how often ARVD/C patients who meet Task Force Criteria actually have a normal ECG. We looked at 145 patients with a documented sustained ventricular arrhythmia who met task force criteria at last follow up. Of these 145 patients, 128 had an abnormal ECG as expected with the most common abnormality being the presence of T-wave inversions. 15 patients had nonspecific ECG findings (not meeting ECG task force criteria) and 2 patients had normal ECGs. Although the majority of patients with arrhythmias have an abnormal ECG, a normal ECG does not preclude the ARVD/C diagnosis. All patients with a normal ECG still met ARVD/C diagnostic criteria. A comprehensive clinical evaluation is necessary to assess arrhythmic risk.

ATRIAL ARRHYTHMIAS PRESENT IN 14% OF PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY

Christian F. Camm, Cynthia A James, Crystal Tichnell, Brittney Murray, Aditya Bhonsale, Anneline S.J.M te Riele, Daniel P Judge, Harikrishna Tandri, and Hugh Calkins

Limited information is available concerning the prevalence of atrial arrhythmias in ARVD/C. This goal of this study was to review the medical records of 249 ARVD/C patients enrolled in the Hopkins Registry to assess the presence of atrial arrhythmias. Thirty-five patients with ARVD/C (14%) experienced one or more types of atrial arrhythmias. Atrial fibrillation was the most common, occurring in 80% of ARVD/C patients with atrial arrhythmias. Older age at last follow up and male gender were associated with development of atrial arrhythmias. Atrial arrhythmias are common in ARVD/C and present at a younger age than the general population.

The Radiological Society of North America held its annual meeting in December 2013 in Chicago, Illinois. Neda Rastegar, MD, a Post-Doctoral Radiology Fellow here at Johns Hopkins, was invited to present the work she has done MRI and ARVD/C.

INCIDENCE OF NON-TASK FORCE CRITERIA FINDINGS BY CMR IN SUBJECTS WITH VARIOUS ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY SCORES

Neda Rastegar, Stefan Zimmerman, Cynthia James, Brittney Murray, Anneline S.J.M. te Riele, Aditya Bhonsale, Crystal Tichnell, Hugh Calkins, Harikrishna Tandri, David A. Bluemke, and Ihab R. Kamel

The goal of this study was to determine the incidence of abnormal MRI findings, in addition to the standard task force criteria, in patients with ARVD/C. Ninety-four patients with an ARVD/C associated desmosomal mutation were included. Thirty patients fulfilled major MRI criteria, 3 fulfilled minor MRI criteria, and the remaining 61 patients did not fulfill any MRI task force criteria. 60% of those that met major MRI criteria had left ventricular fat and/or delayed enhancement. While fat infiltration and delayed enhancement are not considered part of the diagnostic criteria, they may be useful in suggesting more advanced ARVD/C.

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 will also be collected.

How to Join: Contact Crystal 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.

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! Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu

Incidence and Characteristics of Heart Failure Among Patients with ARVD/C

As part of our annual follow up, we will be inviting you to participate in a survey regarding symptoms that may be related to heart failure. We are trying to figure out what types of ARVD/C patients might develop heart failure. Check your email for a link to this survey!

Pregnancy in ARVD/C Patients

We are looking for women with a diagnosis of ARVD/C to participate in our study describing the incidence and course of pregnancy among ARVD/C patients. Surveys have been sent out that ask several questions about your pregnancies, including method of delivery, use of medications, complications, incidence of arrhythmias, etc. If you are a woman diagnosed with ARVD/C and have not received this survey, please contact Crystal at ctichnell@jhmi.edu. Even if you have not had any pregnancies, you are still eligible to participate in this survey.

Effect of Exercise Within An ARVD/C Family

We continue to interview patients about their exercise history to gain a better understanding of the impact exercise has on the progression of ARVD/C. An important next step is taking a look at individuals within a family who carry the same genetic change and how their exercise history has impacted their development or non-development of ARVD/C. Invitations have been sent to eligible individuals and we are hoping to move forward as soon as these interviews are complete.

THANK YOU FOR YOUR PARTICIPATION IN ALL OF THESE IMPORTANT STUDIES!!!

Other Important News

Planning to come to the seminar in May?? Would you like to schedule a consultation with a genetic counselor and electrophysiologist to discuss your diagnosis, management, genetic testing?? Then schedule your appointment now! We will be available for consultations on Friday, May 2nd. These appointments go quickly. Your insurance company will be billed for these services. Contact Crystal @ 410-502-7161 or ctichnell@jhmi.edu to schedule an appointment.

We are working with several groups to characterize the mechanisms of ARVD/C and associated arrhythmia by creating heart muscle cells from blood and skin biopsies. These cells are induced to form "pluripotent stem cells" and then turned into heart muscle cells. We are also continuing to investigate mechanisms of development of cardiomyopathy and arrhythmia in mice with ARVD/C, with a goal of bringing these treatments to people.

The "Feel the Beat" section of the newsletter is dedicated to patient stories. If you would like to share your story in a future newsletter, contact Crystal at ctichnell@jhmi.edu



I have now been living longer with the ARVD diagnosis than without it. I am 44 years old and when I was 20 years old I collapsed during a rowing race and nearly died. At least five times since then I cheated death. I am now onto my third ICD.

For the past 24 years I have learned to live in an entirely new way than my first 20 years. Before my diagnosis, I was an accomplished athlete in peak physical condition, with hopes of making the Olympic rowing team for Canada. I loved to take physical risks; cliff jumping into the water, white-water canoeing, skiing fast and running to exhaustion. I enjoyed pushing my body and testing the limits of my strength and endurance. My heart was my accomplice in adventure.

It has taken many, many years to accept the limitations of my heart. It has been a long journey of frustration, denial, fear and sometimes depression. Not a day goes by when I am not reminded of what I can and cannot do to stay alive. The list of "can't's" are endless; can't row, can't run, can't play tennis, can't ski.....can't, can't can't. It's a four-letter word I would like to banish from my vocabulary.

Here is what I can do. I can walk my dog for miles at a slow pace. I can play golf, fly-fish and kayak gently. I can entertain my friends, dance slowly and travel to major cities. I can cheer for my favorite sports teams and splash around in the water with my niece. I can read, keep a journal, eat healthfully and meditate. I can live with a full and joyful life with ARVD.

Over the years I have grudgingly learned to be more self-compassionate. I have slowly replaced stoicism with vulnerability, and gone from being an extreme type A++ to just an A. I have developed an appreciation for my birthdays, since getting older is a triumph! I have replaced the high of winning a tough tennis match with the satisfaction of making a one-putt. And I have learned that sometimes the bravest thing to do when facing adversity is to care for yourself.

Heather



Raising funds for ARVD/C Research is easy! Just ask the Kraft Family. Christine Kraft and her four children did a little holiday baking for some neighbors on their street this past December. They raised \$240 for their efforts and donated their bake sale proceeds to the Johns Hopkins ARVD/C Research Program. Thank you Kraft Family!

How You Can Help

None of the research by the ARVD/C Program would be possible without the active participation of families affected by ARVD/C. To join our research, the first step is to enroll in the ARVD/C 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/C is a rare disease and not considered a major health hazard. It is critical for patients or families affected by ARVD/C to lobby for increased funding for research on this important condition.

If you are interested in making a financial contribution to the ARVD/C Program, please contact Shannon Wollman, Director of Development for the Johns Hopkins Heart Institute at swollma3@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>

ARVD Program Info

ARVD Program Staff

Hugh Calkins, MD—Director

Harikrishna Tandri, MD—Faculty

Daniel Judge, MD—Faculty

Stuart Russell, MD—Faculty

Theodore Abraham, MD—Faculty

Abhishek Sawant, MD—Post Doctoral Research Fellow

Anneline te Riele, MD—Post Doctoral Research Fellow

Cynthia James, ScM, PhD—Genetic Counselor

Brittney Murray, MS—Genetic Counselor

Crystal Tichnell, MGC—Genetic Counselor

Support Group Info

Looking for a support group?

ARVD support group on Yahoo:

<http://health.groups.yahoo.com/group/mme+tera/>

FACEBOOK Groups:

- **ARVD/C Youth Society** - private group on Facebook (request invite from group admin)

- **Hope for ARVD** - public group

- **The Broken Heart Club - ARVD Edition** - public group

THANK YOU!!!

A special thank you to everyone who has supported the Johns Hopkins ARVD/C Program over the past year. We truly appreciate the generosity of patients and families who have been able to support our program financially. We cannot thank you enough for your continued participation in responding to surveys, sending medical records, providing blood/skin samples, etc. Together, we will continue to solve the mysteries of ARVD/C.

Contact Us

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