

JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

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Summer 2013

2013 ARVD/C Family Seminar

In this Issue...	page
2013 ARVD Family Seminar	1, 2
Make-A-Wish® Recipient	2
Featured Manuscript	2
Heart Rhythm Society Abstracts	3
Hopkins Research Review	4
Research Opportunities	5
Patient Stories	6
Support Group Info	7
Upcoming Fundraisers	7
Make a Donation	7

The 14th Annual ARVD/C Patient and Family Seminar was the best one yet with approximately 170 individuals and family members affected by this condition in attendance. Those that arrived into Baltimore early Friday evening attended a Meet 'n Greet at the Hilton Garden Inn (the new hotel for this year) and enjoyed some hors d'oeuvres and company of old and new friends.

The actual seminar began Saturday morning where participants attended several presentations that addressed various aspects of ARVD/C, including diagnosis, genetics, disease mechanism, catheter ablation, role of exercise, meditation, as well as the psychosocial aspects of living with ARVD/C.

We were excited to have two featured guest speakers this year. Dr. Peter van Tintelen from the *University of Groningen, The Netherlands* presented "Genes, Mutations and ARVC; One Big Family?" Dr. Samuel Sears from East Carolina University returned once again and presented "Confident Living 2013".

Dr. Mario Delmar and his wife, Dr. Marina Cerrone, were guests at our 2012 seminar and enjoyed it so much, they returned to give an update on their current and proposed work on ARVD/C. Dr. Dan Judge also presented our recent collaborative work with Dr. Vincent Chen published in *Nature* earlier this year. After lunch, a large Question & Answer Session was held where attendees could ask questions of Dr. Calkins, Dr. van Tintelen, Dr. Sears, Dr. Judge, and Dr. Delmar. We also offered a meditation workshop to introduce seminar attendees to another option for living well with ARVD/C, as well as a discussion group for attendees in their teens and twenties. In addition, there were several research opportunities throughout the afternoon, including blood draws, Holter patches, and ICD interrogations. We would like to thank everyone who was able to stay and participate in the various research studies. Without you, our program would not exist and we would not be able to improve our understanding of ARVD/C. For those of you who were unable to attend, you were missed and we certainly hope to see you at our 15th Annual Patient and Family Seminar next Spring.



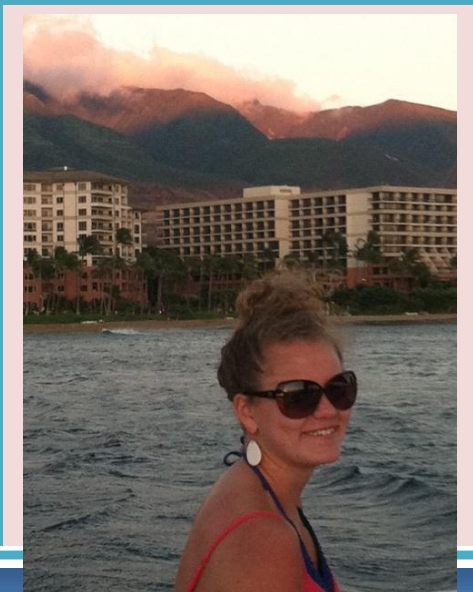
Save the Date!!!

Next Seminar tentatively planned for May 3rd, 2014

ARVD/C Patient chosen for Make-A-Wish®

The Make-A-Wish® Foundation is a non-profit organization founded in the United States that grants wishes to children with life-threatening medical conditions to enrich the human experience with hope, strength, and joy. The foundation grants the wish of a child, on average, every 38 minutes.

Last summer, Callie, an ARVD/C patient, was granted her wish to take a trip to Hawaii. Callie and her family had an amazing time and didn't want to come home!



More Seminar...

Were you unable to attend the ARVD/C Seminar this year? Do you still want to learn the latest about ARVD? Do you wish there was a way you could watch those outstanding presentations? If you answered YES to all of the above, then click the link below to view the 2013 ARVD/C Seminar Presentations. While definitely not a substitute to the overall Family Seminar experience, this year you have the opportunity to view the presentations online. Unfortunately, we can't recreate the special opportunity to interact with the leaders in the field or share personal experiences with other families, a critical aspect to learning how to live with ARVD/C, so start planning now for next year's seminar!

View presentations now at:

<http://tinyurl.com/2013ARVDSeminar>

Featured Manuscript

EXERCISE INCREASES AGE-RELATED PENETRANCE AND ARRHYTHMIC RISK IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY ASSOCIATED DESMOSOMAL MUTATION CARRIERS

Cynthia A. James, ScM, PhD, Aditya Bhonsale, MD, Crystal Tichnell, MGC, Brittney Murray, MS, Stuart D. Russell, MD, Harikrishna Tandri, MD, Ryan J. Tedford, MD, Daniel P. Judge, MD, and Hugh Calkins, MD

We are very excited to share the findings of our exercise interviews that so many of you participated in. Our paper, ***Exercise Increases Age-Related Penetrance and Arrhythmic Risk in Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Associated Desmosomal Mutation Carriers***, describes our findings and has been accepted for publication in the Journal of the American College of Cardiology. The goal of this study was to determine how exercise influences actual disease. While we can't propose that half of those with ARVD/C be allowed to exercise by participating in high level endurance activities and then evaluate them to see how much exercise has advanced their condition, we can try to correlate exercise history with current state of disease. We did just this and found that endurance exercise and frequent exercise increase risk of ventricular arrhythmias, heart failure, and development of ARVD/C among carriers of desmosomal mutations. Restriction from frequent and endurance exercise is extremely important for these patients and modification of exercise at clinical presentation may improve outcomes.

Heart Rhythm Society Abstract Presentations

The 34th Annual Heart Rhythm's Scientific Sessions were held in Denver in May 2013. The research and collaborative efforts of the Johns Hopkins ARVD/C Program were well-represented by several presentations and posters which have been listed/summarized below.

EXERCISE INCREASES PENETRANCE AND ARRHYTHMIC RISK IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY (ARVD/C)

Cynthia A. James, PhD, Aditya Bhonsale, MBBS, Crystal Tichnell, MS, Brittney Murray, MS, Stuart D. Russell, MD, Daniel P. Judge, MD, Harikrishna Tandri, MBBS and Hugh Calkins, MD, FHRS.

This abstract was selected for an oral presentation at the 2013 HRS. It has also been recently published in the Journal of the American College of Cardiology. You can read the summary of this study in the Featured Manuscript Section of the Newsletter.

CARDIAC PHENOTYPE OF ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA

Anneline S.J.M. te Riele, MD, Cynthia A. James, PhD, Binu Philips, MD, Neda Rastegar, MD, Aditya Bhonsale, MD, Judith A. Groeneweg, MD, Brittney Murray, MS, Crystal Tichnell, MGC, Daniel P. Judge, MD, Jeroen F. van der Heijden, MD, Maarten J.M. Cramer, MD, Birgitta K. Velthuis, MD, David A. Bluemke, MD, PhD, Stefan L. Zimmerman, MD, Ihab R. Kamel, MD, Richard N.W. Hauer, MD, Hugh Calkins, MD, and Harikrishna Tandri, MD

The goal of this study was to re-evaluate the MRI findings observed in ARVD/C. The MRIs of 81 mutation positive ARVD/C patients were reviewed for regional abnormalities. The MRI was abnormal in 59 patients with the right ventricle (RV) being abnormal in 93%. Left ventricular (LV) involvement was observed in 53%. Isolated LV involvement was seen in 7%. The most frequent RV abnormality was focal dyskinesia of the basal inferior wall followed by basal anterior wall.

PREVENTION OF SUDDEN CARDIAC DEATH (SCD) IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY (ARVD/C): 92 CASES DIAGNOSED POST-MORTEM

Richa Gupta, BS, Cynthia James, PhD, Crystal Tichnell, MS, Aditya Bhonsale, MBBS, Brittney Murray, MS, Harikrishna Tandri, MBBS, Daniel P. Judge, MD and Hugh Calkins, MD, FHRS.

This poster described 92 cases of ARVD/C diagnosed after sudden cardiac death (SCD) and compared them with 128 gene-positive ARVD/C cases that presented while living. Autopsy reports, medical history, and family history information were collected and reviewed. The average age of SCD was 34.9 years. Men experienced SCD at a younger age than women. SCD occurred most often during daily activity (36%), 23% during exercise, and 14% during sleep. 46% had cardiac symptoms prior to death, syncope being the most common. Only 10% presenting with SCD had a known family history prior to SCD. Following death, 175 first-degree family members were screened resulting in 42 diagnoses.

CARDIAC SARCOIDOSIS MASQUERADING AS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY: EXPERIENCES FROM THE JOHNS HOPKINS ARVD/C REGISTRY

Binu Philips, MD, Srinivasa Madhavan, MD, MSPH, Cynthia James, ScM, PhD, Crystal Tichnell, MGC, Brittney Murray, MS, Saman Nazarian, MD, PhD, Hugh Calkins, MD, Harikrishna Tandri, MD, and Alan Cheng, MD

This poster described clinical features that may aid in the differentiation of ARVD/C and cardiac sarcoidosis which is important because there is considerable overlap in clinical features of these two diseases. Our study evaluated 15 patients with cardiac sarcoidosis who were initially misdiagnosed as ARVD/C and 30 definite ARVD/C patients. ARVD/C patients present earlier and have frequent PVCs. EKG abnormalities including prolonged intervals and second/third degree AV block were commonly seen in cardiac sarcoid patients but not ARVD/C patients. Also, imaging studies commonly showed septal involvement in cardiac sarcoidosis patients.



What's New in ARVD/C Research at Johns Hopkins?

Take a look at some of the exciting research going on at Johns Hopkins!



Maryland Stem Cell Grant



Dr. Leslie Tung, a basic scientist here at Johns Hopkins, has been collaborating with our program over the past several months. As a result, together we applied for and received a three-year grant from the Maryland Stem Cell Research Fund to create tissue models of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy. This project would be the first to model electric currents among cells in three dimensions in the laboratory to mimic the native disease process. Dr. Tung will also stretch these cells to mimic what happens during exercise. This model will also be used to test how arrhythmias in ARVD/C start. Ultimately, by improving our understanding of the electrical currents and mechanisms, we will be able to move forward with improvement in available treatments.

* * * * *

Looking at Arrhythmias

This summer we welcomed Christian Fielder Camm, a medical student from the United Kingdom. Fielder led two projects during his 2 short months with us. His first project focused on the prevalence and types of atrial arrhythmias in patients also diagnosed with ARVD/C. His second project involved using iRhythm's Zio® Patch to evaluate how much variation there is in PVC count from one day to the next during a 7-day period. Many seminar attendees volunteered to wear the patch, along with several members of our team (pictured to the right). We are currently evaluating the data collected and hope to publish results soon. Thank you to everyone who volunteered for this study.

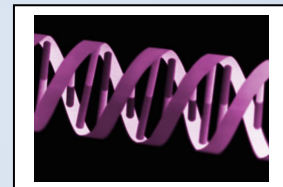


Back: Anneline de Riele, Crystal Tichnell
Front: Cindy James, Brittney Murray, Fielder Camm



* * * * *

Transatlantic Collaboration



We continue a very important collaboration with Drs. Richard Hauer and Peter van Tintelen in the Netherlands and their group of Dutch ARVD/C patients. The goal of this study is to compare the clinical course of over 550 ARVD/C patients and their family members with different genetic findings (no mutation vs mutations, mutations in different genes). Important characteristics that we are evaluating include sustained VT/VF, presence of left ventricular dysfunction, heart failure, and cardiac transplant. Part of this work has been presented at two cardiology meetings; the *Northwestern Cardiovascular Young Investigators' Forum* in September 2012 and the *American Heart Association Scientific Sessions* in November 2012. We are preparing our first manuscript for publication shortly.

* * * * *

Research Opportunities at Johns Hopkins

Clinical and Genetic Investigations of Right Ventricular Dysplasia (ARVD/C Registry)

Who: Children and adults with ARVD/C

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

* * * * *

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/C!

* * * * *

Predictors of ICD Firing in ARVD Patients

Who: People with a definite diagnosis of ARVD/C and an implantable cardioverter defibrillator (ICD)

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.

Contact Crystal (ctichne1@jhmi.edu; 410-502-7161) or Cindy (cjames7@jhmi.edu; 443-287-5985) if you are interested in either of these studies.

Clinical Services at Johns Hopkins

The Johns Hopkins ARVD/C Program also provides a variety of clinical services. We see patients for second opinion consultations to discuss diagnosis and management, genetic counseling and testing, as well as routine ICD management and family member screening. 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. In selected cases we also offer catheter ablation as a treatment for difficult to manage ventricular tachycardia. Appointments with our heart failure specialists, Drs. Stuart Russell, Daniel Judge, and Ryan Tedford can also be arranged. These appointments are billed to your health insurance. To schedule an appointment, contact Crystal at 410-502-7161 or ctichnell@jhmi.edu.

**Don't forget to keep us informed of your most up-to-date contact info!
Please send any changes and updated medical records to Crystal at
ctichnell@jhmi.edu Thank you!**

Feel the Beat Feel the Beat Feel the Beat Feel the Beat

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

*There was a little girl and she had a little curl; Right in the middle of her forehead,
When she was good, she was very, very good; And when she was bad she was horrid.*

My story began almost 3 years ago when I was 34. I had always been healthy, no issues with my heart. In fact, I have had 3 beautiful babies with no complications or issues during any of my pregnancies. No one in my family has had an early-unexpected death. This was totally unexpected.

In the summer of 2010, I was at a family reunion in which I was doing a relay race when my heart began racing and it wouldn't stop. I had to bow out of the race and couldn't continue because my heart was beating so fast, I couldn't catch my breath. Forty-five minutes later there was no change. I left the family reunion to go to the local emergency room. On the way to the hospital I could feel my heart slow down and I honestly thought I could hear the blood in my body go "whoosh" as it slowed. My husband pulled the car over and I ran around the parking lot trying to get my heart to race again because I knew the emergency room staff wouldn't do anything unless it could be seen on an EKG. Nothing happened and I didn't go to the emergency room that day.

I made an appointment with a cardiologist and he said there nothing wrong. In fact, he recommended more magnesium. I wasn't satisfied with that recommendation and I began seeking out additional medical resources to explain what happened. At first I was diagnosed with RVOT. My MRI was fine, my echo showed a bicuspid valve and a slight enlargement of the right ventricle but nothing to worry about, or so I was told. I showed a little period of V-Tac in the stress test, the experts were suspicious something was happening. Four months after the RVOT diagnosis I ended up in the ICU after I was cardio-verted for uncontrollable heart rhythm. At that time, the doctors realized the RVOT was not being managed by medication and an ablation was recommended. The ablation took 3 hours and in the end the doctor tapped me on the shoulder and said one section was ablated. He proceeded to inform me that I didn't have RVOT, instead I had ARVC and I needed an ICD.

It has been one year since being diagnosed with ARVC and it is a year that I will never forget. I have coded 3 times, had an ablation and a catheter ablation, and I have spent 5 days in the ICU for an uncontrollable heart rhythm. I have learned to explain my disease as simply as possible, however people still look at me as if I am going to pass out or die at any second. This is what I don't appreciate. This disease has been a bigger impact on my psychological well being than my physical. Many people give me a lot of attention because they are worried and are fearful of my survival and it is easy for me to get caught up in the worry too. It is comforting to know I have an ICD and the longer I live with this disease the more I realize *"today is not my day to die"*. So far, ARVC has changed my life, but my day to day activities have only been temporarily impacted. The poem listed at in beginning of this story sums up my life at this time. Most days I feel fine. The moments when my heart races unexpectedly, my life is in danger. *"When I am good, I am very, very good, but when I am bad I am horrid."* ~Mollie~

ARVD/C Program Staff



ARVD Program Staff

Hugh Calkins, MD—Director
Harikrishna Tandri, MD—Faculty
Daniel Judge, MD—Faculty
Stuart Russell, MD—Faculty
Theodore Abraham, MD—Faculty
Aditya Bhonsale, MD—PostDoctoral Research Fellow
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Crystal Tichnell, MGC—Genetic Counselor

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Looking for a support group?

ARVD support group on Yahoo:

<http://health.groups.yahoo.com/group/mmetter/>

ARVD/C Youth Society - private group on Facebook
(request an invite from group admin or contact
Brittney at bmurray@jhmi.edu)

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. Continue to lobby for increased funding for research on this important condition. Funding from the NIH and other public organizations remains difficult to obtain. In part, this reflects the fact that ARVD/C is a rare disease and not considered a major health hazard.

If you are interested in making a financial contribution to the ARVD/C Program, please contact Shannon Tower, 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/C 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

7th Annual Golf Tournament

A tribute to Bonnie Milner

Friday, August 9, 2013
8:00 am Shotgun Start
Rocky Point Golf Course
1935 Back River Neck Road
Essex, MD 21221

\$100 per golfer. Includes registration gifts, practice range, lunch, beverages and refreshments on the course, and awards reception. Proceeds to benefit ARVD Research at Johns Hopkins Heart Institute. For info or to be a sponsor, email HealingHeartsMD@yahoo.com. Find "Healing Hearts" on Facebook for more info regarding this organization.



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