

JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

Volume 4, Issue 2

Summer 2014

2014 ARVD/C Family Seminar

The 15th Annual ARVD/C Patient and Family Seminar was another huge success with approximately 180 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 and enjoyed some hors d'oeuvres and company of old and new friends.

We tried something new this year and offered an early morning session for new seminar attendees, newly diagnosed patients, and for anyone else that needed a refresher. The ABC's of ARVD/C was a huge hit and very well attended. Even those that have been living with ARVD/C for years now thought it was helpful. After breakfast, we dove into some exciting presentations that addressed various aspects of ARVD/C, including diagnosis, genetics, disease mechanism, iPS cells, drug discovery, catheter ablation and role of exercise.

We were thrilled to have two featured speakers this year. Dr. Cristina Basso, MD, PhD from Padova, Italy presented "Historical Perspective on ARVC – Padua Experience" and "Athletes, Arrhythmias, and Sudden Death". Dr. Jeff Saffitz, MD, PhD returning from Beth Israel Deaconess Medical Center presented "Drug Discovery for ARVC".

After lunch, a large Question & Answer Session was held where attendees could ask questions of Dr. Basso, Dr. Saffitz, Dr. Tandri, and Dr. Calkins. We also offered Discussion Group Workshops for the Youth Group (under 30ish), the over 30 affected group, as well as for family members and support persons. In addition, there were research opportunities throughout the afternoon, including blood draws 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 thrive and we would not be able to solve the mysteries of ARVD/C. For those of you who were unable to attend, you were missed and we certainly hope to see you at our 16th Annual Patient and Family Seminar next Spring.

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Did you miss the ARVD/C Seminar this year? Do you still want to learn the latest about ARVD/C? Then click the link below to view the 2014 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>

More Seminar...



Read what people are saying about the ARVD/C Seminar...

Best seminar ever! Very Informative.

Fantastic Day! Thanks so much for a day of learning and support!

Well done! Balanced...Understandable – but not “dumbed down”.

I like to hear from those that have ARVC. Very good seminar!

Q & A is excellent because you learn about other patients’ concerns. Research was very efficient – BRAVO!

Excellent – outstanding...leaves me hopeful and very grateful!

Great seminar...The information presented this year was extremely informative and very encouraging.

This was an excellent conference. I feel privileged to be invited to attend this conference and to hear the latest research. The experts are excited and engaging in presenting their information.

Under 30 youth group was very helpful.

The seminars keep getting more and more amazing and very knowledgeable!

Best one I’ve been to!

Both my wife and I thoroughly enjoyed the whole weekend.

The breadth of the morning session was extensive, from history to current research, and very much appreciated. It was encouraging to hear about the collaboration that occurs throughout the research world of ARVD/C.

You don’t want to miss the next one...

Save the Date!!!

**Next Seminar tentatively
planned for May 2nd,
2015**



Hugh Calkins, Cristina Basso, Hari Tandri, Jeff Saffitz

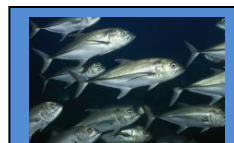
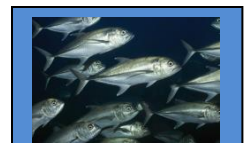
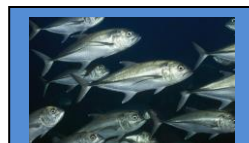
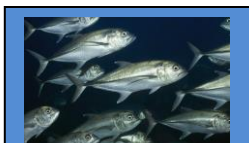
Photos courtesy of Kiele Binsted

Featured Manuscript

IDENTIFICATION OF A NEW MODULATOR OF THE INTERCALATED DISC IN A ZEBRAFISH MODEL OF ARRHYTHMOGENIC CARDIOMYOPATHY

Angeliki Asimaki, Sudhir Kapoor, Eva Plovie, Anne Karin Arndt, Edward Adams, ZhenZhen Liu, Cynthia A. James, Daniel Judge, Hugh Calkins, Jared Churko, Joseph C. Wu, Calum A. MacRae, Andre G Kleber, Jeffrey E. Saffitz. *Sci Transl Med* 6, 240ra74 (2014)

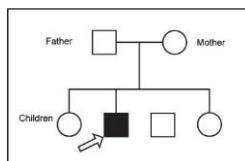
It was an honor and privilege to have Dr. Saffitz join us again this year at our May Seminar to share more of his zebrafish work! Dr. Saffitz and his team have developed a zebrafish model of Arrhythmogenic Cardiomyopathy to better understand the mechanisms behind ARVD/C. They then began screening thousands of potential chemicals and discovered a potential drug, SB216763 that suppressed the disease expression in the zebrafish model. SB216763 demonstrated the ability to prevent or reverse features of ARVD/C in zebrafish, as well as in rat models. They have observed strong evidence of a trafficking abnormality in ARVD/C that accounts for a variety of features of the disease. Additional work is being done with SB216763 and iPSCs from human patients with ARVD/C.



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. We can arrange cardiac testing including Echocardiograms, Cardiac MRIs, Stress tests, Electrocardiograms, Signal Averaged Electrocardiograms, and 24 hour Holter monitors. More invasive testing, such as Electrophysiology Studies and Mapping, Right ventriculograms, and Biopsies can also be arranged when necessary.

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. We see all of our patients for genetic counseling to discuss the diagnosis, the psychosocial impact of living with ARVD/C and with an ICD, as well as to discuss the benefits and limitations of appropriate genetic testing. 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.



Mycvmd.com

Heart Rhythm Society Abstract Presentations

The 35th Annual Heart Rhythm's Scientific Sessions were held in San Francisco in May 2014. 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 INTENSITY DURING YOUTH MODIFIED BY DESMOSOMAL MUTATION STATUS PREDICTS AGE OF PRESENTATION IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA

Abhishek Sawant, MD, MPH, Cynthia A. James, PhD, Anneline te Riele, MD, Crystal Tichnell, MGC, Brittney Murray, MS, Ryan Tedford, MD, Daniel P. Judge, MD, Harikrishna Tandri, MD and Hugh Calkins, MD, FHRS.

This abstract was selected for an oral presentation at the 2014 HRS meeting. The goal of this study was to evaluate the impact of exercise among both carriers and non-carriers of desmosomal mutations. 106 patients meeting task force criteria were interviewed (58 mutation carriers) about their activity level beginning at age 10. Patients who presented before age 25 had higher rates of participation in "Class C" sports (per 36th Bethesda Conference) between ages 10-24. Mutation carriers were more likely to present before age 25. Non-carriers were more likely to be Class C athletes before presentation. Mutation non-carriers may require higher intensity exercise to develop disease.

ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY IN A LARGE TRANSATLANTIC PATIENT COHORT

Judith A Groeneweg, Aditya Bhonsale, Cynthia James, Jeroen van der Heijden, Brittney Murray, Crystal Tichnell, Anneline te Riele, Abhishek Sawant, Pieter Doevendans, Jan DH Jongbloed, Harikrishna Tandri, J Peter van Tintelen, Daniel P. Judge, Toon A van Veen, Dennis Dooijes, Arthur A Wilde, Richard Hauer, Hugh Calkins.

This abstract was selected for an oral presentation at the 2014 HRS meeting. The clinical and genetic characteristics of ARVD/C patients meeting 2010 Task Force Criteria and their family members from the US/Dutch Registries were assessed in this study. 395 index patients and 578 family members were included. Mutations (mostly PKP2 43%) were identified in 61% of index patients. ARVD/C was diagnosed in 179 (31%) relatives. Familial ARVD/C occurred in 54% (83/153) of mutation positive vs 34% (20/59) mutation negative families. Among 920 subjects presenting alive, 30 (3%) died, 332 (36%) had a sustained arrhythmia, 49 (5%) experienced heart failure, and 20 (2%) require cardiac transplantation.

YIELD OF SERIAL EVALUATION IN AT-RISK FAMILY MEMBERS OF ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PATIENTS

Anneline te Riele, MD, Cynthia James, PhD, Neda Rastegar, MD, Aditya Bhonsale, MD, Brittney Murray, MS, Crystal Tichnell, MS, Daniel Judge, MD, David Bluemke, MD, PhD, Stefan Zimmerman, MD, Ihab Kamel, MD, PhD, Hugh Calkins and Harikrishna Tandri, MD

In this study, 33 unaffected relatives of 22 ARVD/C probands underwent serial comprehensive cardiac screening. None of the patients fulfilled diagnosis criteria at first evaluation. Over a follow-up period of more than 4 years, our study showed that 1) disease progression in at-risk relatives of ARVD/C probands occurred in 1/3 of subjects; and 2) electrical abnormalities preceded detectable structural changes.

OUTCOMES OF FIRST-LINE EPICARDIAL ABLATION OF VENTRICULAR TACHYCARDIA IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY

Binu Philips, MD, Anneline S.J.J te Riele, Abhishek Sawant, MD, Vishnupriya Karedy, Cynthia James, ScM, PhD, Brittney Murray, MS, Crystal Tichnell, MGC, Bina Kassamali, Saman Nazarian, MD, PhD, Daniel P. Judge, MD, Hugh Calkins, MD and Harikrishna Tandri, MD.

This goal of this study was to compare the outcome of first-line epicardial ablation strategy to epicardial ablation after failing endocardial ablation of VT. Twenty-eight consecutive patients with definite ARVD/C were included in the study. Fourteen underwent primary epicardial ablation (Group 1) and were compared with 14 patients who underwent epicardial following a failed endocardial procedure (Group 2). Group 1 showed a trend towards longer VT free survival compared to Group 2. Overall epicardial ablation was associated with a significant reduction of the VT burden in both groups. No complications occurred except for acute pericarditis in one (4%) patient.

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

GENETICS, MECHANISMS AND CLINICAL PHENOTYPES OF ARRHYTHMOGENIC CARDIOMYOPATHY

Johns Hopkins ARVD/C Program is participating in a newly funded, multicenter, NIH sponsored grant as an enrolling center.

We are looking for subjects diagnosed with ARVD/C who participated in the previous NIH-sponsored ARVD/C study to re-enroll as well as their family members. We are also looking for newly diagnosed subjects to enroll, along with their family members.

In this study we are trying to find the gene(s) that are responsible for ARVD/C, and to see how the gene(s) affect the onset, the course and the severity of the disease in one individual and/or in a family. Participation will involve sending us your records, yearly follow up, ECGs, 24 hour Holter monitoring, Signal averaged ECG, 6-minute walk test, and blood donation.

If you are interested and want to learn more about your participation and eligibility, please contact Crystal Tichnell, MGC at 410-502-7161 or ctichnell@jhmi.edu.

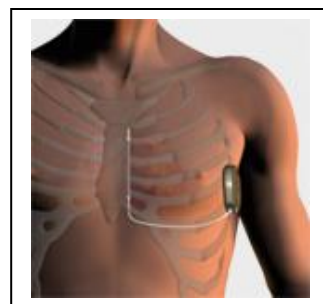


Sub-Study of the Clinical and Genetic Investigations of ARVD/C – PREGNANCY IN ARVD/C

We are looking for women who have been diagnosed with ARVD/C, meeting the task force criteria, prior to becoming pregnant. If you think you fall into this category and would like to participate in this study describing pregnancy in ARVD/C, please contact Crystal at ctichnell@jhmi.edu.

The S-ICD – Is it for me?

This is a question that has been asked a lot recently. Stay tuned to learn more about a possible upcoming research opportunity to help us figure out which ARVD/C patients might benefit from the new S-ICD. If you already have an S-ICD system, please enroll in our registry if you haven't done so already. Email Crystal at ctichnell@jhmi.edu.



Bostonscientific.com

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.

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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!

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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.

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

In this edition, I'd like to share two videos that some ARVD/C patients have participated in to help spread awareness of ARVD/C.

Inspiring Kids: Nick's story

<http://www.youtube.com/watch?v=7-27xgC9AUg&list=UUdNccTRUvei5nXGhz971NJg>



* * * * *



Ryan's story (although the title of the video says Hypertrophic Cardiomyopathy, Ryan is living with ARVD/C)

https://www.youtube.com/watch?v=7p3yYQywCNI&feature=em-share_video_user

* * * * *

Zach's Story

Everything changed on March 13, 2012, during a pickup basketball game before my first senior year varsity baseball practice. I jumped up to get a rebound, and when I landed something didn't feel right. I ran off the court, and passed out in the corner of the gym. My friend Jarrett Riker ran to find our coach, and brought him to the gym.

My coach said that as soon as he opened the door and saw me laying face down in the corner, he knew there was something seriously wrong. He said my eyes were closed, my face was turning purple, and I was not breathing. He yelled to one of the guys to go grab the AED from the gymnasium office. He then rolled me over on my back, I took a gasping breath, and color started returning to my face. The AED was not needed. An ambulance was called, and took me to the nearby hospital.

Doctors initially suspected that I had simply fainted, and released me from the E.R. Over the next few days, I continued to feel lightheaded and had chest pains. My parents took me back to the Emergency Room. After many tests were administered, the doctor there, consulted with a Neurosurgeon at a larger hospital, and it was determined that I needed to set up a consultation with that Neurosurgeon as soon as possible. Once meeting with him, and being put through several tests, it was determined that I suffered from Arrhythmogenic Right Ventricular Dysplasia (ARVD).

I was scheduled for surgery to implant the ICD on April 26th, but became quite sick before then, and my surgeon pushed my surgery date up to Friday, April 13th.

Since then, it has been very difficult for me to not be able to play all of the team sports I once enjoyed so much. Fortunately, I am able to play golf, and try to do so as much as possible. There are times when I overexert myself, and I know that I need to take a rest.



There are so many times I ask "Why Me?", but I know that is one question I will never have answered.

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.

While we continue to apply for funding to support our various research projects, we rely heavily on the generosity of families to financially support this program.

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 443-287-7383. 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 CONTINUED SUPPORT!!!

ARVD/C 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

Looking for a support group?

ARVD support group on Yahoo:

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

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

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!

Contact Us

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