

JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

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2012 ARVD Family Seminar Summary

In this Issue...	page
2012 ARVD Family Seminar	1
Heart Rhythm Society Abstracts	3
Support Group Info	2
Hopkins Research Review	4
Research Opportunities	5
Patient Stories	5,6
Upcoming Fundraisers	7
Make a Donation	7
ARVD Program Contact Info	7

The 13th Annual ARVD Patient and Family Seminar was another huge success with approximately 185 individuals and family members affected by this condition in attendance. Those that arrived into Baltimore early Friday evening attended a Smoothie Social at the hotel and were able to meet up with old friends and make new ones as well. The actual seminar began Saturday morning where participants attended several presentations that addressed various aspects of ARVD, including diagnosis, genetics, disease mechanism, catheter ablation, cardiac transplant, yoga and nutrition. We were excited to have two guest speakers this year. Dr. Mario Delmar from NYU Langone Medical Center presented "The Science of ARVD/C: What Have We Learned?" Dr. Stefanie Toise from Hospital of Saint Raphael shared her work in investigating the efficacy of adapted yoga in managing the anxiety and depression in patients with ICDs. After lunch, a large Question & Answer Session was held where attendees could ask questions of Dr. Calkins and Dr. Delmar. We were also able to offer an informational Nutrition workshop as well as a Yoga Workshop. In addition, there were several research opportunities throughout the afternoon. 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 solve the mysteries of ARVD. For those of you who were unable to attend, you were missed and we certainly hope to see you at our 14th Annual Patient and Family Seminar next Spring.

Save the Date!!!

Next Seminar tentatively planned for May 4th, 2013

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 2012 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/2012ARVDSeminar>

Support Group Info

ARVD/C Youth Society

We are excited to announce the beginnings of a "Youth" focused support group. There has been a growing discussion amongst the younger generation of individuals with ARVD/C to establish a support group to address the new and unique challenges in terms of school, college, work, starting a family, etc, while juggling a new diagnosis or genetic test result for themselves or family member.

This group is open to anyone under the age of 30ish who is either diagnosed with ARVD/C, carries an ARVD/C mutation, and/or has a family member with ARVD/C that would like to connect with others sharing similar experiences.

The goals and specific format of the group are up for discussion based on the needs of the group members, but have included a discussion forum, fundraising, advocacy, and organizing fun events around the seminar. A small group of individuals met at the 2012 seminar and have started a private Facebook page as a way to facilitate communication.

The goal is to allow free communication between youth members. If your youth family member is interested, please ask them to sign up via the brief informational page at <https://www.surveymonkey.com/s/ARVDCYouthGroup> in order to be added to the email list and join the private Facebook group. You can ask to be removed from the group at any time. Feel free to contact Brittney at bmurray@jhmi.edu with any questions about this group.

International ARVD Family Support Network

Established in April of 1999, this is a private and confidential self-help support community. Membership is composed of people who have either been diagnosed with ARVD/C, or had one of their family members diagnosed with it and is composed of almost all age groups. The purpose of the group is to support one another and to share information about ARVD/C. The group meets via the Internet through a private "Yahoo Groups" website and/or list serve (email communications) which is directed and moderated by a volunteer staff.

To join the group, you or one of your family members must have received a diagnosis of ARVD/C. Potential group members are briefly interviewed prior to group admission. Members must agree to respect the private/confidential nature of the group. Because this is a private group, membership is subject to the discretion of group leadership. In order to join the support group, you will need to take a moment to "Sign Up" at Yahoo Groups first. Once you are signed up at Yahoo Groups, you can apply to join the "mmettera" group. To begin this process, go to <http://health.groups.yahoo.com/group/mmetter/>.

Note: These support groups are NOT monitored by Johns Hopkins ARVD Program Staff.

Heart Rhythm Society Abstract Presentations

The 33rd Annual Heart Rhythm's Scientific Sessions were held in Boston in May 2012. The research and collaborative efforts of the Johns Hopkins ARVD Program was represented by several presentations and posters which have been summarized below.

OUTCOMES OF EPICARDIAL CATHETER ABLATION OF VENTRICULAR TACHYCARDIA AMONG PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA

Binu Philips, Srinivasa Madhavan, Cynthia A. James, Crystal Tichnell, Brittney Murray, MS, Aditya Bhonsale, Hugh Calkins, and Harikrishna Tandri

The purpose of the study was to assess the efficacy of epicardial ablation of ventricular tachycardia in patients with ARVD/C. The study population consisted of 27 patients with ARVD who underwent 29 epicardial VT ablations at 11 centers in the US. Of the 27 patients, 20 had previously failed endocardial ablation procedures. One year after the epicardial procedure, 61% of patients were free of VT. Two years later, 48% were free of VT and five years later, 48% were free of VT. Eleven of 17 patients on antiarrhythmic medication prior to the procedure remained on antiarrhythmic medication at follow up. In summary, about half of patients have recurrence of VT after epicardial catheter ablation.

SIGNAL-AVERAGED ELECTROCARDIOGRAPHY REVEALS ARRHYTHMIC RISK AMONG ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY ASSOCIATED DESMOSOMAL MUTATION CARRIERS

Aditya Bhonsale, Cynthia A James, Crystal Tichnell, Brittney Murray, Binu Philips, Srinivasa Madhavan, Harikrishna Tandri, Daniel P. Judge, and Hugh Calkins

The goal of this study was to investigate the role of Signal Averaged ECG (SAECG) in determining ventricular arrhythmia risk among patients with an associated desmosomal mutation. Data was collected on 133 patients known to have a desmosomal mutation (83% PKP-2). Over a period of 36 ± 15 years, 55 (41%) patients experienced an arrhythmic outcome. Filtered QRS duration (fQRS), root mean square voltage in the last 40 ms (RMS), and duration of signal <40 μ V (LAS) were significantly different between those with and without arrhythmic outcome with the higher quartile cutoffs associated with more arrhythmic outcomes. SAECG parameters can assist in risk stratification.

A SCORING SYSTEM FOR DISTINGUISHING VENTRICULAR ARRHYTHMIAS IN PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY FROM RIGHT VENTRICULAR OUTFLOW TRACT TACHYCARDIA

Kurt Hoffmayer, MD; Prashant Bhawe, MD; Gregory M. Marcus, MD, MAS; Crystal Tichnell, MGC; Hugh Calkins, MD; Nitish Badhwar, MBBS; Edward Gerstenfeld, MD; and Melvin Scheinman, MD

The goal of this study was to determine if an ECG scoring system using morphology of VT or PVCs with a left bundle inferior axis pattern could be used to differentiate between ARVD/C and right ventricular outflow tract tachycardia (RVOT-VT). The scoring system looked at normal sinus rhythm anterior T wave inversions V1-V3 (3 points), QRS duration in lead I ≥ 120 msec (2 points), QRS notching (2 points), and precordial transition V5 or later (1 point). A score of 5 or greater was able to distinguish ARVD/C from RVOT-VT 93% of the time.

Dan Judge spoke in a session called "Risk stratification in the cardiomyopathies" and discussed how to use genetic test results as well as clinical factors to help determine who has a particularly high risk of developing (dangerous?) ventricular arrhythmia. Hugh Calkins led a session called "ARVD/C Three Decades of Progress" that also included ARVD Experts geneticist Peter van Tintelen, cardiologist William McKenna, and pathologist Cristina Basso. Dr. Calkins discussed "Management considerations for ARVD/C: Who should get an ICD and what about exercise." Cindy James and other genetic counselors presented a session called "Navigating Genetic Testing tips from the Experts." She explained how to work with families to organize genetic testing once a mutation has been found in the family.

Hopkins Research Review

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

Dr. Judge's lab recently developed a mouse with a DSG2 mutation that shows clear signs of ARVD/C. Over the next few years he will use this mouse to better understand how mutations cause ARVD/C and to test potential ARVD/C treatments.

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We are collaborating with Dr. Richard Hauer of the University of Utrecht in the Netherlands to determine how mutations are associated with different clinical features of ARVD/C. We have combined genetic and registry data of North American and Dutch patients so that we can better study what features of ARVD/C correspond to particular mutations. This will allow us to provide better advice and treatment.

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We are part of an exciting new NIH-sponsored study to look for genes associated with Mendelian (inherited) conditions. This study uses new rapid sequencing to look for genetic variation in the genes of people with a variety of inherited conditions. We have been approved to enroll families with ARVD/C who have had desmosomal genetic testing but have had no mutation found yet.

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We have begun to study the pathophysiology of ARVD/C – that is we are investigating what happens at a cellular level to cause the loss of heart cell muscles, arrhythmias, and heart failure found in ARVD/C. We are working with a number of other physicians and scientists across the country. In collaboration with Dr. Jeffrey Saffitz and his team at Beth Israel Deaconess Medical Center in Boston, we have learned that regardless of which mutation a person with ARVD/C has, there is a large reduction in the amount of plakoglobin present in heart muscle cells. Also in collaboration with Dr. Saffitz we are investigating whether the death of heart muscle cells in ARVD/C is related to inflammation.

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We are in collaboration with a couple of groups that are studying ARVD/C induced pluripotent stem cell lines. These cell lines are grown from taking skin or blood and transforming stem cells in the skin/blood to heart cells for research study.

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One focus of our program continues to be improving MR imaging (MRI) for ARVD/C. This program is currently led by Dr. Hari Tandri, Dr. Stefan Zimmerman, and Dr. Ihab Kamel. We are investigating new techniques to closely investigate MRI features of people with ARVD/C-associated desmosome mutations.

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

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.

***For those of you who are part of the ARVD Registry thank you for your ongoing willingness to share your experiences with us. This year we'll be asking questions about your exercise history when we check in to see how you are doing. We hope you can spend some time on the phone telling us about the sports/activities you have participated in over the years.**

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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!

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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.

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.

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

Well my name is Kiele Binsted, I just turned 25, and I'm from Greenville, SC. So I have a lot to say in a little amount of time so I'm going to jump right in. It wasn't until Early last year I was officially diagnosed with ARVD, but I've experienced tachycardia and other symptoms since the 6th grade. So in 2009 I graduated college and became a seasonal worker. I was living the good life, a worry free life. I travelled and worked in all kinds of different cities and resorts. In the fall of 2010, I just finished working my third summer in Glacier National Park, Montana, when a couple friends and I moved to South Lake Tahoe to work at Kirkwood Ski Resort. But three days after our arrival, my plans suddenly changed. We had just signed a lease for a new apartment and had only been unpacking our cars for about 30 minutes when I went into VT. Since this has been happening to me since 6th grade, I was very familiar with it, and usually, I could make it go away by sitting down and resting. This time was different. This time I didn't pass out and literally had to grasp for every breath. I was taken to the hospital and given a couple shocks. Later, I had been told I was minutes away from full on Cardiac Arrest. I was eventually transferred to Reno, NV, which is

where the closest EP to Tahoe was. I stayed a month in Reno, half the time in the hospital getting my first ablation, and my ICD implanted, and the other half with my parents in a hotel room, who were kind enough to fly out from SC.

Now to catch you up to speed in lieu of this incident, like I said before I've been suffering from Tachycardia since the 6th grade. During my sports especially basketball my episodes of Tachycardia caused me to blackout where I would still have control of my body, but I would lose complete eyesight. The same symptoms continued all through college until my sophomore year in 2006, when the disease started to progress causing me to pass out.

I did see a Cardiologist for about 6 years but b/c all my tests came out negative he was convinced that I was dehydrated, and so I was convinced even after my Tahoe incident, I thought it was merely dehydration. I was in complete denial that something more serious was affecting me.

Then, in the beginning of 2010, my symptoms started to become very sporadic. I was living in Portland at the time and passed out twice in two months. The first time was out of nowhere as I was walking to work, and a month later, I passed out again after giving a four hour swim lesson in a heated pool. I ended up having a severe concussion and a month of vertigo. Then two months after my last syncope episode in Portland, I decided to move back to Glacier for one more summer. The first night in town I had a bout with Tachycardia that led to syncope. But instead of returning to sinus rhythm, like I usually had, this time when I awoke I was still in VTach for a couple hours. That was the only time I experienced syncope that summer, although I was still plagued by many episodes of Tachycardia, usually when I was playing Frisbee or basketball, but thankfully never when I was out in the middle of the woods climbing mountains. So after the season ended in Glacier, in September of 2010, that's when my friends and I moved to Tahoe, and that's when things got chaotic. After my month stay in Reno I was stable enough to fly back to South Carolina. Shortly after getting home I kept going into VT so I had my ablation in January of last year. It helped some but not enough. Because a couple months after the ablation, in March, my defibrillator went off for the first time. It shocked me four times. I handled it well, and carried on as if nothing had happened. It wasn't until a month later that things took a turn for the worst, when my defibrillator went off for the second time. It unexpectedly shocked me twice while I was in bed about to fall asleep. It frightened me to think that I was not safe even in my own bed, that I could be shocked anytime or anywhere.

Since nothing obvious, triggered the shocks, doctors finally discovered an underlying genetic kidney disorder that only aggravated my ARVD. Basically, my kidneys don't hold on to my potassium or magnesium, causing critically low levels that produce arrhythmias.

After my last round of shocks, I had extreme anxiety. I couldn't sleep in my own bed for a couple weeks, I couldn't sleep at all for that matter, and when I could I suffered from phantom shocks, so I didn't even want to fall asleep. I thought every pvc was going to turn into another shock and sometimes I still think that. I started to analyze each and every heartbeat. I had no peace of mind. That's when Brittney and the Doctors here decided it was a good time for me to have an epicardial ablation. Hearing that was music to my ears. The procedure was done last year in the first week of July, and it was an all-day affair, it was very successful so as soon as it was over Dr. Tandri immediately took me off my anti-arrhythmics even though I was unbelievably apprehensive about it, but I had complete trust in him. Afterwards the procedure my sleep returned, my energy returned, and most importantly my heart has been stable. So I am very thankful for all their hard work.

It wasn't until a couple of months ago that I was really able to get active again. Not only was my anxiety still high, but I was plagued by a number of different health issues. It seemed like my whole body wanted to breakdown. I even had to get my gallbladder removed just one week after the last ablation. But for the most part, my health has been stable lately, so I've started taking trips back out west to see my friends, I've started attending concerts again but most importantly I've started Cardiac Rehab. I love Rehab, nurses monitor me while I'm in a relaxed environment which makes me feel at ease when I exercise. I now am aware of my limits, and I am comfortable with an elevated heart rate. So because of Rehab, my anxiety is not controlling me anymore and I've gained back some of that peace of mind I lost. But I will say there's still not a single day that goes by when I don't think about getting shocked.

Like many of you, the other biggest struggle I've had, besides the anxiety, has been the absence of physical activity. I've been a competitive athlete as far back as I can remember, whatever the sport, it didn't matter, even my jobs were physically demanding. It's been quite the dramatic lifestyle change. It's hard to know that I can't go on a long run, or get in a good swim workout, or play a pick-up game of Basketball or Frisbee. But thinking about that only gets me down so I'm constantly reminding myself that "Life could always be worse." I remind myself that there are plenty of people in the world who are suffering much greater than I am and somehow that always manages to put life back in perspective for me. It helps me realize I still got it pretty good. Even if I can't participate in all the fun and games I once knew, I can still read and write and take pictures, I can go for walks, spend more time with the family, do Yoga, go out with friends, travel, create things, further my cooking skills, make art, learn to play an instrument. The possibilities are endless. I've even picked up Golf and returned to one of my favorite activities, Disc Golf. Just because we're prohibited from strenuous activity that doesn't mean we are prohibited from discovering new talents and new skills that we may have never once known. Life is always changing, whether for the good or the bad, so we must learn to adjust and adapt at what life hands us. Life's too short to spend time and energy dwelling on what I can't do and what I don't have, so I take that time to focus on the things I can do, the things I have, the things that are sure to bring me happiness in life, because life is just too short not to be embraced. Now, this bracelet that I always wear, is one that I found in my grandmother's belongings after she passed away. She battled breast cancer for many years, and the bracelet says to Celebrate Life. I never take it off because that's what she did during her struggles and that's what I'm doing. You only live once and it's worth the celebration. So adjust, adapt, embrace the situation, and carry on, because, remember, it could always be worse.

ARVD Program Staff

Our website has a
NEW LOOK!!!

Check it out @
www.ARVD.com



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Crystal Tichnell, MGC—Genetic Counselor

What would you like to see in the next
newsletter?

Email Crystal with your ideas: ctichnell@jhmi.edu

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

6th Annual Golf Tournament

A tribute to Bonnie Milner

Friday, August 3, 2012
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. Visit www.HealingHeartsMD.com for info regarding this organization and events.

