

JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA PROGRAM

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2011 ARVD Family Seminar Summary

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The 12th Annual ARVD Patient and Family Seminar was another huge success with approximately 175 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, catheter ablation, and cardiac transplant. We were excited to have two guest speakers this year. Dr. Jeffrey Saffitz from Beth Israel Deaconess Medical Center in Boston presented "New Insights into the Diagnosis and Mechanisms of Disease." Dr. Samuel Sears was thrilled to return for a third time to discuss "The Amazing Race: You and Your Family and ARVD." After lunch, a large Question & Answer Session was held where attendees could ask questions of Dr. Calkins, Dr. Saffitz, and Dr. Sears. 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 13th Annual Patient and Family Seminar next Spring.

Were you unable to attend the ARVD 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 2011 ARVD Seminar Presentations. While definitely not a substitute to the overall Family Seminar event, this year you have the opportunity to view the lectures 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, so start planning now for next year's seminar!

<http://webcast.jhu.edu/mediasite/Catalog/pages/catalog.aspx?catalogId=49d3ff66-75ad-4aaf-8855-78f697b0d8a0>
or <http://tinyurl.com/2011ARVDSeminar>

*The Johns Hopkins ARVD Program would like to announce our 13th Annual ARVD Patient and Family Seminar to be held on **Saturday, April 28th, 2012**. Hope to see you there! Registration will open in December.*



??? Questions from the Seminar ???

Q: Once you are diagnosed with ARVD, what kind of follow up treatment should you expect from your doctors? Should you have an electrophysiologist follow you?

A: In general, a patient diagnosed with ARVD should be evaluated with ECG, Holter monitoring, and Echo on a yearly basis to monitor for progression. In addition, regular device checks are also recommended. An electrophysiologist is a cardiologist who has special training in heart rhythm disorders and is usually more familiar with the arrhythmia aspects of ARVD.

Q: What are the recommendations for ARVD patients regarding exercise?

A: At this point in time, it is recommended that patients diagnosed with ARVD avoid competitive athletics and limit their exercise to low intensity activities. Research continues in this area to determine more definitive recommendations.

Q: What is the recommended testing interval for siblings of a patient with ARVD?

A: In general, it is recommended that first degree relatives of someone diagnosed with ARVD be evaluated every 2-3 years with ECG, signal averaged ECG, exercise stress test, echo and/or MRI, and 24 hour Holter monitor. Additional testing may be recommended based on the results of these tests. The interval between screenings may also vary depending on symptoms, results of prior testing, family history, and genetic test results.

Q: I hear others are taking medications - I don't take any for my heart. What kinds of medications are typical for patients with ARVD?

A: The specific types of medications that may be prescribed vary from patient to patient. However, some of the common medications include beta blockers and ACE-inhibitors.

Research Opportunities at Johns Hopkins

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!

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.

Heart Rhythm Society Abstract Presentations

The Heart Rhythm Society is the international leader in science, education and advocacy for cardiac arrhythmia professionals and patients, and the primary information resource on heart rhythm disorders. Its mission is to improve the care of patients by promoting research, education and optimal health care policies and standards. The Heart Rhythm Scientific Sessions were held in San Francisco in May 2011. The research and collaborative efforts of the Johns Hopkins ARVD Program was represented by 9 abstract presentations which have been summarized below.

INCIDENCE AND PREDICTORS OF APPROPRIATE ICD THERAPY IN PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA (ARVD) UNDERGOING ICD IMPLANTATION FOR PRIMARY PREVENTION - *Young Investigator Award*

Aditya Bhonsale, MD, Cynthia A. James, PhD, Crystal Tichnell, MS, Brittney Murray, MS, Dmitri Gagarin, MD, Darshan Dalal, MD, Binu Philips, MD, Ryan Tedford, MD, Harikrishna Tandri, MD, Daniel P. Judge, MD and Hugh Calkins, MD

The purpose of this study was to define the incidence and predictors of appropriate ICD therapy in ARVD patients following placement of an ICD for primary prevention (no sustained arrhythmias yet). 84 patients with a diagnosis of ARVD who underwent ICD placement were described. Over a mean follow up of 4.7 +3.4 years, 48% of patients had an appropriate shock. Inducibility on EP study and presence of nonsustained VT are strong predictors of appropriate ICD therapy in ARVD patients receiving an ICD for primary prevention.

PSYCHOSOCIAL ADJUSTMENT TO LIFE WITH AN ICD AMONG ARVD PATIENTS

Cynthia A James, PhD, Crystal Tichnell, MS, Brittney Murray, MS, Hugh Calkins, MD and Samuel Sears, PhD.

The goal of this study was to compare patients with ARVD/C on standardized measures of general and disease-specific adjustment. Eighty-six adults completed a set of questionnaires measuring ICD-specific anxiety, device acceptance, anxiety and depression, and functional capacity. While overall acceptance of an ICD appears to be high, anxiety is elevated, particularly among those who are young and who have experienced a device discharge. Clinical attention to screening and appropriately referring ARVD/C patients for treatment of anxiety is recommended.

MODELING ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA (ARVD) USING PATIENT-SPECIFIC INDUCED PLURIPOTENT STEM CELLS

Changsung Kim, PhD, Joseph E. Marine, MD, Hugh Calkins, MD, Daniel P Judge, MD and H-S Vincent Chen, MD, PhD

This study describes how several induced pluripotent stem cell (iPSC) lines were successfully generated from fibroblasts of a patient with known PKP2 mutations. These ARVD iPSC-heart muscle cells were able to beat, as well as show features of ARVD including arrhythmias, fatty infiltration and cell death. These cells may be used in the future to explore treatment strategies for patients with ARVD.

ONE MUTATION FITS ALL: PHOSPHOLAMBAN R14DEL UNDERLIES BOTH ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC) AND DILATED CARDIOMYOPATHY (DCM)

Paul A van der Zwaag, MD, Angeliki Asimaki, PhD, Jan D Jongbloed, PhD, Maarten P. van den Berg, MD, PhD, Ans C. Wiesfeld, MD, PhD, Moniek G. Cox, MD, Laura T. van Lochem, MD, Rudolf A. de Boer, MD, PhD, Karin Y. van Spaendonck-Zwarts, MD, Isabelle C. van Gelder, MD, PhD, Daniel P. Judge, MD, Hugh Calkins, MD, Albert J. Suurmeijer, MD, PhD, Richard N. Hauer, MD, PhD, Jeffrey E. Saffitz, MD, PhD, Arthur A. Wilde, MD, PhD and J. Peter van Tintelen, MD, PhD.

This abstract describes the identification of a genetic change in the phospholamban (PLN) gene that has been observed in both ARVC and DCM. The specific change, R14del, was identified in 31 of 240 (13%) patients with DCM and 12 of 97 (12%) patients with ARVC.

RE-EVALUATION OF SIGNAL AVERAGED ELECTROCARDIOGRAPHY IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY: THE ROLE OF GENDER IN IDENTIFYING OPTIMAL CUT-OFFS

Rahul Jain, MD, MPH, Raoul Manalac, BS, Crystal Tichnell, MS, Cynthia James, PhD, Rohit Jain, MD, Hari Tandri, MD, Theodore Abraham, MD, Stuart D. Russell, MD, Ronald D. Berger, MD, PhD, Gordon F. Tomaselli, MD, Hugh Calkins, MD and Darshan Dalal, MD, PhD.

The purpose of this study was to examine the diagnostic utility of SAECG using a large group of patients evaluated for ARVD/C. SAECG parameters differed significantly between men and women. As a result, this study proposed gender-specific cut-offs for SAECG parameters, which offer better diagnostic utility in evaluating patients for ARVD.

MULTICENTER EFFICACY OF ENDOCARDIAL CATHETER ABLATION OF VENTRICULAR TACHYCARDIA IN PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA

Binu Philips, MD, Srinivasa Madhavan, MD, MPH, Dmitri Gagarin, MD, Aditya Bhonsale, MD, Cynthia James, PhD, Brittney Murray, MS, Crystal Tichnell, MS, Darshan Dalal, MD, PhD, Saman Nazarian, MD, Stuart D. Russell, MD, Theodore Abraham, MD, Daniel P Judge, MD, Harikrishna Tandri, MD and Hugh Calkins

This study examined 60 patients with ARVD who underwent a total of 115 catheter ablation procedures of VT. The majority of patients had at least 2 procedures. During a mean follow up period of 17 months, VT recurred after 101 of the total 115 procedures. Approximately 73% of the recurrences were within the first year. Repeat procedures had a significantly better 1 year arrhythmia free survival. Note: we have found that newer ablation procedures work better and people with recent ablations have a considerably more favorable outcome.

CARDIAC TRANSPLANTATION IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA

Ryan J. Tedford, MD, Cynthia James, PhD, Daniel P. Judge, MD, Crystal Tichnell, MS, Brittney Murray, MS, Aditya Bhonsale, MD, MBBS, Binu Phillips, MD, Dmitri Gagarin, MD, Harikrishna Tandri, MD, Darshan Dalal, MD, Hugh Calkins, MD and Stuart D. Russell, MD.

This study describes 21 patients diagnosed with ARVD who were listed for cardiac transplant. 20 patients underwent successful transplant. Pre-transplant, 15 patients met criteria for definite ARVD. 3 patients were excluded after assessment of the explanted heart revealed an alternate diagnosis. An associated disease-causing mutation was identified in 7 of 15 patients. Heart failure was the most common reason for transplant. Biventricular dysfunction was also common.

MYOCARDIAL EXPRESSION AND ELEVATED CIRCULATING LEVELS OF INFLAMMATORY CYTOKINES IN ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY

Angeliki Asimaki, MD, PhD, Harikrishna Tandri, MD, Elisabeth R. Duffy, MSc, William J. McKenna, MD, Hugh Calkins, MD, Shiva Gautam, PhD, Daniel G. Remick, MD and Jeffrey E. Saffitz, MD, PhD.

The analysis of heart biopsies from ARVC patients revealed significantly increased levels of pro-inflammatory cytokines, proteins that serve as messengers between cells. Levels of a specific anti-inflammatory cytokine were significantly depressed in patients with ARVD. Analysis of inflammatory biomarkers in future studies should allow better risk stratification and correlation with arrhythmias.

PENETRANCE OF DESMOSOMAL MUTATIONS IN FAMILIAL ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA (ARVD)

Kunal Sanghavi, MS, Aditya Bhonsale, MD, Crystal Tichnell, MS, Brittney Murray, MS, Harikrishna Tandri, MD, Hugh Calkins, MD, Daniel P. Judge, MD and Cynthia A. James, PhD

This study ascertains the penetrance of desmosomal mutations and the influence of the new 2010 ARVD diagnostic criteria. Detailed clinical information from 28 families with an ARVD-associated desmosomal mutation was obtained. Variable expression and penetrance was observed among these families.

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.

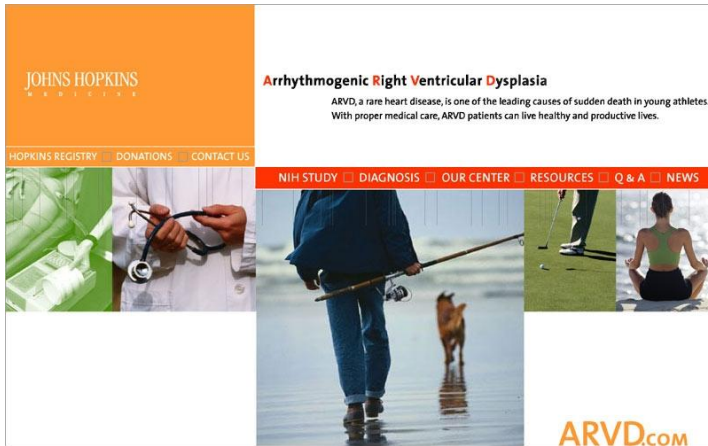
*For those of you who are part of the ARVD Registry thank you for your ongoing willingness to share your experiences with us. This year we'll be asking questions about your exercise history when we check in to see how you are doing. We hope you can spend some time on the phone telling us about the sports/activities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

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

I was diagnosed with ARVD when I was 21 years old. I went from playing in a collegiate tennis match to having an ICD implanted 24 hours later. There were clues along the way that something was wrong, but what seemingly healthy 21 year old thinks they actually have a heart condition? I had to adapt quickly to my new life as I started getting shocked within 4 months of receiving the implant. Since then I have been shocked 13 times, some of them one after another. I have been shocked for many different reasons ranging from dancing, chasing after a dog that got loose, to doing absolutely nothing at all. I quickly learned that being active in athletics was no longer even a choice. You do a lot of soul searching when the life you once knew gets flipped upside down. What I came to find was that I still have a life and I intend to live it to the fullest. Every day is a gift. I would be lying if I said it wasn't tough at times. I am now 28 years old, and within my years of knowing I have ARVD, I have had an ICD Replacement, 4 heart ablations, and an ICD Lead Extraction and Replacement just to name a few things. The reason why I am still left with a positive and optimistic attitude is because we have the most amazing team on our side. I do not think I would be who I am with this disease if it weren't for the doctors and genetic counselors we have fighting our fight. Personally, I have an amazing support system with a brother who shares my fate and parents who attend every conference and are in the waiting room for ever surgery. I have a husband who married me knowing it would be through sickness and health. I do have a heart condition, but I also have a beautiful life.... *Rebecca Sieg*, living with ARVD

ARVD Program Staff



ARVD Program Staff

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What would you like to see in the next newsletter?

Email Crystal with your ideas
ctichnell@jhmi.edu

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

5th Annual Golf Tournament

Friday, August 12, 2011

8:00 am Shotgun Start

Rocky Point Golf Course

1935 Back River Neck Road

Essex, MD 21221

\$90 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.

