

# JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

Volume 7, Issue 1

Winter 2017

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

Are you ready to take on 2017??? From all of us here at the ARVD/C Program, we hope you and your families have a wonderful Holiday Season. We wish you good health, happiness, and success in the coming year and always! This past year the ARVD/C Program has continued working with collaborators from around the world. We've wrapped up some projects and launched some new ones so once again we'll be looking for your help.

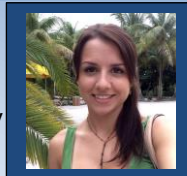
Please know that your continued support both financially as well as through your research participation does not go unnoticed. Thank you for volunteering your records, your time, and your ideas. Advances that have been made in the field of ARVD/C are all because of your dedication. We truly value this partnership and look forward to improving our understanding of ARVD/C together. We cannot thank you enough!

We are excited to announce our Annual Patient and Family Seminar this Spring and hope to see many of you there. Details are enclosed. Please do not hesitate to contact our program with any questions or concerns.

~ The ARVD/C Program

## Welcome New Staff

Julia Agafonova recently joined the program in October as our Research Assistant. She moved from Russia to the United States in 2006 and graduated from the State University of New York College at Brockport with a degree in Psychology. Following her passion in research, she worked on several projects in New York and developed an interest in genetics. Julia will be managing many of the regulatory aspects of our research, as well as working very closely with our abundance of data. Ultimately, Julia plans to earn her degree in Genetic Counseling and continue helping patients affected by genetic disorders.



## Planning to come to the seminar in April??

Would you like to schedule a consultation with the ARVD/C Team to discuss your diagnosis, management, and/or genetic testing??

→ Then request your appointment **NOW!** ←

Appointments will be available:  
Thursday, April 27<sup>th</sup> (Dr. Tandri)  
Friday, April 28<sup>th</sup> (Dr. Calkins)  
Monday, May 1<sup>st</sup> (Dr. Calkins)

These appointments go quickly! Your insurance company will be billed for these services. Contact Crystal @ 410-502-7161 or [ctichnell@jhmi.edu](mailto:ctichnell@jhmi.edu) to request an appointment.

## Congratulations to Aditya Bhonsale,

a past ARVC fellow, who received the ARVC Clinical Research Prize at the Third International Zurich ARVC Symposium in Switzerland this past September 2016 for his work on the *Impact of Genotype on Clinical Course in ARVC mutation carriers*. This work was described in the Winter 2015 Newsletter. The prize is part of the Zurich ARVC Program established in 2011 to increase awareness of this challenging disease and promote in-depth clinical and basic research in this field at an international level.



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# 18<sup>th</sup> 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 share their experiences with ARVD/C this year: Professor Firat Duru, MD, Dr. Ali J. Marian, MD and Dr. Samuel Sears, PhD.

Professor Duru will be joining us from Zurich, Switzerland where he is Director of the Cardiac Pacing and Electrophysiology Division, Department of Cardiology at the University Hospital, Zurich. He and his colleagues developed a Center of Excellence for ARVC in Zurich in 2011 and have started a large clinical registry of ARVC patients and their family members.



We are truly excited to have Dr. Samuel Sears, cardiac psychologist from East Carolina University, Greenville, North Carolina, back again this year. Dr. Sears is known for his work in understanding psychosocial adjustment of patients living with ICDs, response to ICD treatment, and shock acceptance strategies to name a few.

Dr. Marian is the Director of the Center for Cardiovascular Genetic Research at The Brown Foundation Institute of Molecular Medicine at The University of Texas, Health Science Center at Houston. He is recognized nationally and internationally for his research achievements and expertise in genetics of cardiomyopathies, as well as understanding the molecular pathogenesis of ARVD/C.



In addition, we will have several Johns Hopkins faculty members present this year: Dr. Hugh Calkins – management of ARVD/C, Dr. Hari Tandri – catheter ablation and sympathectomy, Dr. Dan Judge – Genetics, and Brittney Murray, genetic counselor, overview of ARVD/C.

After lunch, there will be an opportunity to participate in small group discussions led by Dr. Sears, as well as opportunities to participate in research.

Make plans to come into Baltimore early to attend a reception at the Hilton Garden Inn Friday evening. Once again, we have an exciting and packed agenda so we look forward to seeing many of you in April! Please register early! There is no registration fee for this seminar. All are welcome!

**WHEN:** Saturday, April 29<sup>th</sup>, 2017 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  
1800 Orleans Street,  
Baltimore, Maryland 21287

**REGISTRATION:** ALL participants must register! It is also helpful to list names of family members that will be attending with you so we can determine appropriate research opportunities. Register online by April 15<sup>th</sup>.

<http://tinyurl.com/2017ARVCSeminar>

# 18<sup>th</sup> Annual ARVD/C Patient and Family Seminar

Presented by  
The Johns Hopkins ARVD/C Program

## **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 \$152/night plus tax (single/double) until March 29th, 2017 or as long as they are available.

Reservations can be made at Hilton Garden Inn through one of the ways below:

- 1.) Hilton Garden Inn Central Reservations Line at **1-888-429-7482**. Request **Johns Hopkins ARVD 2017 Group rate** / special group code as follows
- 2.) Booking on the web at [www.baltimoreinnerharbor.stayhgi.com](http://www.baltimoreinnerharbor.stayhgi.com) and use special code **ARVD** in the box labeled **Group/Convention code**.
- 3.) Direct Booking Link: <http://group.hiltongardeninn.com/JHUARVD2017>

The hotel front desk can be reached at 410-234-0065. Check-in 3pm / Check-out 12pm. Self-Parking is available at a rate of \$23 and valet \$35 per day.

## **TRAVEL TIPS**

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

Two buses 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 at 5:00pm to return to the Hilton Garden Inn.

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

## **PARKING AT THE SEMINAR**

If you are not taking the bus from the hotel, parking is available at your own expense (max \$12) in the Orleans Street Garage. There is a bridge that connects the garage to the main level of Sheikh Zayed Tower.

## **SPECIAL EVENT**

Join us for a Meet 'n Greet Reception, **7:00-9:00pm**, on **Friday, April 28<sup>th</sup>, 2017** in the Great American Grill at the Hilton Garden Inn. H'ors deuvres will be served. Please register for this event when you register for the seminar or contact Crystal.

## **CLINIC CONSULTATIONS**

Dr. Hugh Calkins and Dr. Hari Tandri and the genetic counselors will be available Thursday, April 27<sup>th</sup>, Friday, April 28<sup>th</sup> and Monday, May 1<sup>st</sup> 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.

## **RESEARCH**

Research opportunities will be available on Saturday afternoon and will include blood drawing, ICD interrogations, etc. **We must have your medical records ahead of time** to determine if you are eligible for research.

## Abstracts and Presentations

The Johns Hopkins ARVD/C Program was well-represented this year at the American Heart Association Scientific Sessions, held November 12-16 in New Orleans, Louisiana. Program Director, Dr. Hugh Calkins, joined ARVD/C experts from around the world for session “Arrhythmogenic Cardiomyopathy: An Update.” In his presentation, “AC and Exercise”, Dr. Calkins shared what we have learned about the role of exercise in the cause and course of ARVD/C. This well-attended session also included past ARVD/C Family Seminar guests Drs. Jeffrey Saffitz, Peter van Tintelen, Gaetano Thiene, and Bill McKenna. Dr. Hari Tandri shared his novel approaches to “Therapy for ARVC in 2016” in a session focused on emerging treatments for ventricular arrhythmias in inherited arrhythmia disorders. Meanwhile, Drs. Dan Judge and Cindy James shared our research and clinical experiences in genetics and genetic counseling. In sessions focused on using large-scale DNA sequencing to improve clinical management Cindy discussed “The Downsides of Big Data: Incidental findings from Comprehensive Genetic Testing.” In a later session moderated by Dr. Judge, she reminded participants of the “Psychological Implications of Incidental Cardiac Genetic Findings for Patients, Families, and Clinicians.”

Our trip to the AHA also gave us the chance to meet with our research partners to discuss preliminary findings and plan new studies. We had the pleasure of meeting with 2016 ARVD/C Family seminar speaker Professor Andre LaGerche, in town from Australia, to plan studies to improve ARVD/C diagnosis and perform novel MRI assessments of exercise on the hearts of people at-risk for developing ARVD/C. Dr. Sam Sears, expert in cardiac psychology and frequent ARVD/C seminar speaker met with us to analyze results of recent questionnaire studies. Finally, we continued our important collaborations with the Dutch ARVC program, planning a variety of ongoing studies to improve clinical management of ARVD/C.

We are excited to continue our collaborations around the world in understanding ARVD/C.



## 2017 Full Member JEMF Awardee

The Jane Engelberg Memorial Fellowship (JEMF) is a highly prestigious award of the National Society of Genetic Counselors. Congratulations to Brittney and the ARVD/C genetic counseling team for being this year’s JEMF Full Member awardee for a project entitled *Outcomes of Genetic Counseling for Arrhythmogenic Cardiomyopathy: A comparison of face-to-face and tele-genetic counseling*.

Increasingly, it has become apparent not only within the profession, but also to the scientific and general population, that the demand for genetic counseling is increasing. Access is limited, however, by both the size and geographic location of the current genetic counseling workforce. Therefore, tele-genetic counseling is an attractive option to increase access. This is of particular importance in cardiovascular genetic counseling, in which genetic testing is often high stakes in managing sudden death risk, however the workforce is limited, with fewer than 200 practicing cardiovascular genetic counselors.



We have been fortunate to receive donor funding to establish a tele-genetic counseling program within our clinic of individuals with inherited arrhythmogenic cardiomyopathy. This provides an important, time-dependent, opportunity to determine whether tele-genetic counseling is non-inferior to traditional face-to-face genetic counseling in achieving three key cardiac genetic counseling outcomes selected based on the Reciprocal Engagement Model: reducing cardiac-specific anxiety, increasing disease-specific genetic understanding, and enhancing patient empowerment. Thus we proposed a prospective observational study of individuals attending genetic counseling for an arrhythmogenic cardiomyopathy indication. Data will be collected by questionnaire 2 weeks preceding and 2 weeks following genetic counseling. In addition to comparing outcomes of face-to-face and tele-genetic counseling, we will assess how genetic counseling outcomes are mediated by the strength of the genetic counselor / client alliance in each setting. Results of the study will provide some of the first evidence of genetic counseling outcomes in cardiology clinics, and also contribute outcome data of alternative methods to expand genetic counseling services.

**Congratulations Brittney!**

## Clinical Services at Johns Hopkins

The Johns Hopkins ARVD/C Program provides a variety of clinical services. We see patients for second opinion consultations to discuss diagnosis and management, genetic counseling and testing, routine ICD management and family member screening. We can also arrange concurrent cardiac testing.

Patients are seen in consultation with Dr. Hugh Calkins or Dr. Hari Tandri and our clinical genetic counselor, Brittney Murray, 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.

As we mentioned in the summer, we have taken steps to expand our clinical services via telemedicine for those unable to travel to Baltimore. This service currently consists of the genetic counseling/genetic testing appointment only. We hope to be able to offer second opinion consultations with our physicians in the future. Telemedicine appointments are currently not billable to your insurance company and require payment out of pocket. This service may not be available in certain states. To schedule an appointment, contact Crystal.

The world's only specialized Pediatric ARVD/C Clinic began in the Fall 2015. This is a monthly clinic staffed by a genetic counselor, Brittney Murray, and our pediatric ARVD/C specialist, Dr. Jane Crosson, pediatric electrophysiologist. We will offer second opinions/consults for both patients possibly affected and also cardiac testing associated with screening for family history of ARVD/C. Contact Crystal for information regarding upcoming dates for this specialty clinic.

Crystal Tichnell, MGC      [ctichnell@jhmi.edu](mailto:ctichnell@jhmi.edu)      410-502-7161

### Hopkins ARVD/C Webinar

In case you missed it, in October 2016, we presented a Hopkins sponsored webinar on ARVD/C. Those who logged on to watch were able to submit questions throughout the presentation for Dr. Calkins and Brittney to discuss. View the presentation by clicking on the link below:

<https://webcast.jhu.edu/Mediasite/Play/0da359dcec804705a6de54db9e3c122a1d>

## Research Opportunities at Johns Hopkins

### Relative Safety, Efficacy, and Patient Satisfaction of Standard ICDs versus the Sub-Cutaneous ICD

**Who can participate?** Patients diagnosed with ARVD/C and have:

- ✓ S-ICD implanted
- ✓ Transvenous ICD implanted after January 2013

**What do I have to do?** Contact Crystal ([ctichnell@jhmi.edu](mailto:ctichnell@jhmi.edu)).

You will need to sign a consent form, send us your medical records, and complete online questionnaires.

**Do I have to travel to Johns Hopkins?** No

**UPDATE:** We have the best research participants! 98% compliance in completing the online questionnaires. THANK YOU!!! We are still looking for additional participants so email Crystal to discuss your eligibility and enrollment.

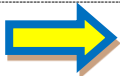
Sponsored by Boston Scientific  
PI: Hugh Calkins, MD  
Johns Hopkins IRB NA\_00042471 (*Predictors of Implantable Cardioverter Defibrillator (ICD) Firing in Right Ventricular Dysplasia*)

### **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 will also be collected.

**How to Join:** Contact Crystal at 410.502.7161 or [ctichnell@jhmi.edu](mailto: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/C who have had an epicardial ablation to join our Registry. Help us discover how this new technique affects the course of ARVD/C! Contact Crystal at 410-502-7161 or [ctichnell@jhmi.edu](mailto:ctichnell@jhmi.edu)

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

### **UPDATE: How Does Family History Influence Psychosocial Adaptation to Inherited Cardiomyopathies?**

The goal of this project, sponsored by the National Society of Genetic Counselors 2015 Jane Engelberg Memorial Fellowship, is to determine how family history influences the overall well-being of individuals diagnosed with an inherited cardiomyopathy and their at-risk family members. We are now working on phase 2 which involves telephone interviews with a subset of participants. We are busy analyzing all of the data and will report back soon!





### Structural Progression in Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy

Thomas P Mast, Cynthia A James, Hugh Calkins, Arco Teske, Crystal Tichnell, Brittney Murray, Peter Loh, Stuart D Russell, Birgitta K Velthuis, Daniel P Judge, Dennis Dooijes, Ryan Tedford, Jeroen F van der Heijden, Harikrishna Tandri, Richard N. Hauer, Theodore P Abraham, Pieter A Doevendans, Anneline S J M te Riele, Maarten J Cramer

We had the pleasure of hosting Thomas Mast, a PhD student from the University Medical Center Utrecht, the Netherlands for a visit last year. Dr. Mast's PhD project focuses on using novel echocardiography techniques to diagnose early ARVD/C. During his visit, we leveraged his expertise in echocardiography to develop a project to characterize structural progression in ARVD/C. We are thrilled to report the paper resulting from this project was recently accepted by the medical journal *JAMA Cardiology*. We are proud of taking this next step of our fruitful Dutch/North American Collaboration and hope it will provide physicians with important information in how they assess progression in their ARVC patients.

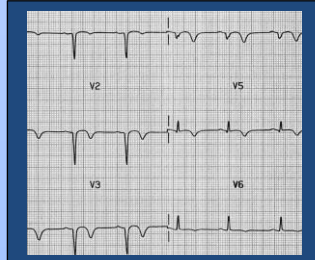
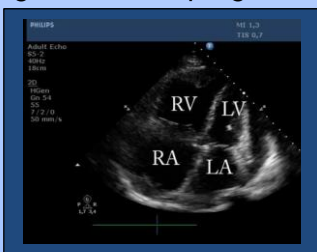
The full article will be freely available shortly. (Check our website for updates with the link to the paper.)

Briefly, the purpose of this project was three-fold: 1) To define the extent of structural progression, 2) To identify predictors of structural progression, and 3) To determine the association between structural progression and ECG changes in patients with ARVD.

The first and last available echocardiograms from 85 patients diagnosed with ARVC and meeting task force criteria were assessed for structural changes, specifically progression. ECGs at baseline and last follow up were also collected and assessed for the presence/absence of diagnostic criteria, as well as the development of new criteria at follow up. Results of this study provided definitive evidence of the structural disease progression in patients with ARVD/C. During a period of 6 years, we observed a decline in RV systolic function and an increase in RV size. LV progression was also observed. Structural progression was usually slow and there was considerable variability among patients indicating the multiple factors that must play a role. Interestingly, the age of onset does not appear to predict the rate of progression.

Importantly, both ECG and Echocardiography are required to monitor the overall disease progression in ARVD/C.

Additional studies are needed to further look at the role of medications, as well as exercise, in how they might influence progression of ARVD/C.



### Your Support of the Johns Hopkins ARVD/C Program Ensures Our Success

As a charitable, tax-exempt organization, Johns Hopkins Medicine relies on donations to make a difference in the lives of our patients. Supporters of Dr. Calkins, Dr. Tandri, and their team of experts in the ARVD/C Program, become part of our mission to provide exceptional personalized care and to find better ways to diagnose and treat our patients. Here are some of the many ways that you can help:

#### Make a Donation

Donations of all sizes, whether they're one-time or recurring, make a difference and can be made online at [www.arvd.com](http://www.arvd.com), by phone at 443-287-7382, or by mail (information listed below). There are a variety of ways to make a gift to support our efforts in the ARVD/C Program:

- Make an outright gift of cash or securities
- Become a monthly donor
- Give in honor or in memory of a loved one
- Give through IRA's, wills and trusts
- Leverage matching gifts through your workplace

#### Fundraising

There are many opportunities to become personally involved in raising awareness and much-needed funds on behalf of the Johns Hopkins ARVD/C Program:

- Create an online giving page and leverage social media
- Ask friends to make contributions in lieu of gifts
- Host your own event or auction
- Plan a fundraising event in your community or school
- Contribute a portion of your company's sales

#### The Johns Hopkins Heart and Vascular Institute Development Office is here to help!

We welcome your questions, concerns, ideas, and feedback. Please contact **Adrienne Rose**, Senior Associate Director of Development, at 443-287-7382 or [arose25@jhmi.edu](mailto:arose25@jhmi.edu), for more information.

#### Gifts by Mail:

The Johns Hopkins Heart and Vascular Institute  
600 North Wolfe Street, Blalock 536B  
Baltimore, MD 21287

**\*Indicate the "ARVD/C Program" on the memo line**



## ARVD/C Program Info

### ARVD Program Staff

Hugh Calkins, MD—Director  
Harikrishna Tandri, MD—Faculty  
Daniel Judge, MD—Faculty  
Stuart Russell, MD—Faculty  
Ryan Tedford, MD—Faculty  
Theodore Abraham, MD—Faculty  
Gabriela Orgeron, 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  
Bryana Rivers—Genetic Counselor Assistant  
Julia Agafonova—Research Assistant

**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 via Fax: 410-502-9148 or email [ctichnell@jhmi.edu](mailto:ctichnell@jhmi.edu) You can also mail records as well. Thank you!**

### Contact Us

Johns Hopkins Hospital  
600 North Wolfe Street, Balock 545  
Baltimore, Maryland 21287  
P: 410-502-7161, F: 410-502-9148  
Website: [www.ARVD.com](http://www.ARVD.com)

## Support Group Info

### Looking for a support group?

#### ARVD support group on Google:

Search for "ARVD ARVC Support Group" on google groups. Any issues joining this group, email Bob at [ralla52@yahoo.com](mailto:ralla52@yahoo.com)

#### FACEBOOK Groups (private):

- ARVD/C Youth Society
- Hope for ARVD
- The Broken Heart Club – ARVD Edition –
- ARVC can't stop me from...

#### ARVD/C Mentor Program:

Get matched with an ARVD/C mentor!

Connect with a mentor who has navigated the challenges of life with ARVD/C and receive:

- Support
- Connection
- Understanding
- Strategies for Thriving with ARVD/C.



Contact Nancy Bogle at [nbstjohn@gmail.com](mailto:nbstjohn@gmail.com) for more information and visit [ARVDHEARTANDSOUL.org](http://ARVDHEARTANDSOUL.org)