JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

Volume 5, Issue 1 **Winter 2015**

* ** ***!!!HAPPY NEW YEAR!!!*** ** *

WELCOME 2015!!! From all of us here at the ARVD/C Program, we hope you and your families have had a wonderful Holiday Season and are looking forward to a fresh new start to the New Year! This past year the ARVD/C Program has continued working with collaborators from around the world. This year we welcomed two students, George and Anke, who both worked on very important projects during their stay. Find out more about them and their projects later on in this newsletter.

Thank you for always responding to emails and requests for information throughout the years. Your dedication to partnering with us is critical in order to solve the mysteries of ARVD/C. We are excited to announce our Annual Patient and Family Seminar this Spring and hope to see many of you there. Please do not hesitate to contact our program with any questions or concerns. We look forward to seeing many of you at our upcoming Patient & Family Seminar in May 2015. Details are enclosed. Best wishes to each of you for a Healthy 2015!

Clinical Services at the ARVD/C Program

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, as well as routine ICD management and family member screening. We can arrange cardiac testing and catheter ablation procedures as necessary. All patients are seen in consultation with Dr. Hugh Calkins or Dr. Hari Tandri and one of the genetic counselors to discuss test results, diagnosis, family history, genetic testing, psychosocial impact of living with ARVD/C and guidance regarding further management. 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.

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Planning to come to the seminar in May??

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! ←

We will be available for consultations on Friday, May 1st and Monday, May 4th. These appointments go quickly. Your insurance company will be billed for these services. Contact Crystal @ 410-502-7161 or ctichnell@jhmi.edu to request an appointment.

16th 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 this year: Professor Richard Hauer from the Interuniversity Cardiology Institute of The Netherlands will share the results of our collaborative efforts; Dr. Vincent Chen from University of California, San Diego will share his stem cell work in ARVC; and Dr. Samuel Sears from East Carolina University in Greenville, North Carolina will discuss the psychosocial impact of living with ARVC and how to "Keep Calm and Carry On". Back this year – The ABC's of ARVD/C – to be presented as an early morning session for new patients and new seminar attendees as a basic intro to understanding ARVD/C. This session is definitely worth the early arrival. After lunch, there will be an opportunity to ask questions in a "*Question and Answer Session with the Experts*", as well as an "Under 30" Discussion Group and 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 May! Please register early!

WHEN: **Saturday, May 2nd, 2015 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 at 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 15th.

https://www.surveymonkey.com/s/2015ARVCSeminar

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 $145/night plus tax (single/double) until March 30th, 2015 or as long as they are available. Call 1-877-STAY-HGI or 1-877-782-9444 and mention the "ARVD Group room block rate" to receive the special rate. Check-in 3pm / Check-out 12pm. Hotel front desk phone is 410-234-0065. Self-Parking is available at a rate of $23 and valet $32 per day.
www.baltimoreinnerharbor.hgi.com
Reservations can also be made online using this link: http://hiltongardeninn.hilton.com/en/gi/groups/personalized/B/BWIIHGI-RVD-20150429/index.jhtml?WT.mc_id=POG

TRAVEL TIPS

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

A bus 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, May 1st, 2015** in the Great American Grill at the Hilton Garden Inn. H'ors deurves will be served. Please register for this event when you register for the seminar or contact Crystal.

CLINIC CONSULTATIONS

Dr. Hugh Calkins and the genetic counselors will be available both Friday, May 1st and Monday, May 4th 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.

QUESTIONS

Contact Crystal Tichnell, MGC at 410-502-7161 or ctichnell@jhmi.edu

JOHNS HOPKINS
MEDICINE

Abstracts and Presentations

The American Heart Association's Scientific Sessions is the leading annual convention for scientists and healthcare professionals devoted to the science of cardiovascular diseases and stroke, and the caring of patients suffering from these diseases. Its mission is to build healthier lives, free of cardiovascular diseases and stroke. The American Heart Association Scientific Sessions were held in Chicago, Illinois in Nov 2014. Nine abstracts (3 presentations and 6 posters) were presented at this year's conference and three are summarized below.

SYSTEMIC APPROACH TO ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY FAMILY SCREENING IN A LARGE TRANSATLANTIC COHORT

Anneline S.J.M. te Riele, Cynthia A. James, Abhishek Sawant, Judith Groeneweg, Kai Kammers, Brittney Murray, Crystal Tichnell, Jeroen F van der Heijden, Daniel P Judge, J. Peter van Tintelen, Richard N Hauer, Hugh Calkins, Harikrishna Tandri

Relatives of patients with ARVD/C are at increased risk of sudden death, but heterogeneous disease expression complicates their risk assessment. We sought to 1) determine predictors of manifest ARVD/C; and 2) optimize arrhythmic risk stratification in at-risk relatives. Records of 345 relatives from 140 families were reviewed and ascertained as per 2010 Task Force Criteria. One-third of at-risk relatives had overt ARVD/C. Siblings are at the highest risk of disease. A combination of symptoms, being a sibling, pathogenic mutation in the family, and female gender allows for accurate prediction of overt ARVD/C.

EXERCISE INFLUENCES PENETRANCE AND OUTCOMES IN FAMILY MEMBERS OF ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY (ARVD/C) PATIENTS CARRYING A PATHOGENIC DESMOSOMAL MUTATION

Abhishek Sawant, Brittney Murray, Crystal Tichnell, Aditya Bhonsale, Anneline S.J.M. te Riele, Stuart D. Russell, Harikrishna Tandri, Ryan J. Tedford, Daniel P. Judge, Hugh Calkins, and Cynthia A. James

Endurance exercise is associated with adverse arrhythmic outcomes and development of heart failure in ARVD/C-associated desmosomal mutation carriers. However, studies comparing exercise participation in family members with desmosomal mutations are lacking. Exercise interviews were performed focusing on duration, intensity and timing among ten families with 37 patients (9 probands, 28 family members) carrying a PKP2 mutation. Mutation carrier family members of probands who are endurance athletes have increased penetrance, worse arrhythmic outcome and greater structural changes than family members who are not

PEDIATRIC-ONSET DISEASE DOES NOT HERALD ADVERSE CLINICAL COURSE IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY

Anneline S.J.M. te Riele, Cynthia A. James, Abhishek Sawant, Brittney Murray, Crystal Tichnell, Ryan Tedford, Jane Crosson, Daniel P Judge, Hugh Calkins, Harikrishna Tandri

In most genetic cardiomyopathies, early disease onset heralds adverse clinical outcome. However, the clinical attributes and disease course in pediatric cases with ARVD/C are largely unknown. The goal of this study was to delineate and compare the clinical characteristics, genetics, and outcomes of pediatric-onset ARVD/C. Among 316 definite ARVD/C cases, 43 (14%) were diagnosed prior to age 18 years. Pediatric patients are disproportionately mutation carriers, but not more likely to carry multiple mutations. There were no other differences observed. Over more than 6 years follow-up, arrhythmic, heart failure, and mortality outcomes are the same in pediatric and adult ARVD/C patients.

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

GENETICS, MECHANISMS AND CLINICAL PHENOTYPES OF ARRHYTHMOGENIC CARDIOMYOPATHY

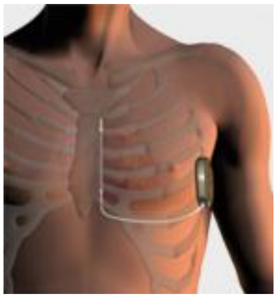
Johns Hopkins ARVD/C Program is participating in a newly funded, multicenter, NIH-sponsored grant as an enrolling center. We are looking for subjects diagnosed with ARVD/C who participated in the previous NIH-sponsored ARVD/C study to re-enroll, as well as their family members. We are also looking for new patients diagnosed with ARVD/C 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. In-person visits are required.

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.

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

Cindy James, ScM, PhD has been awarded the Jane Engelberg Memorial Fellowship Award by the National Society of Genetic Counselors for her project entitled "*How does family history influence psychosocial adaptation in individuals with inherited cardiomyopathies and their at-risk family members?*" Over the next two years, we will be administering questionnaires to those enrolled in the Johns Hopkins Hypertrophic Cardiomyopathy and ARVC research registries, as well as conducting telephone interviews with a subset of patients. Study results will 1) provide an evidence base for adaptation to inherited cardiomyopathy among North American patients and family members, 2) identify risk factors for poor adaptation, and 3) potentially confirm and explain the protective effect of family history.

Be on the lookout for an email to participate in this study if you are already enrolled in our ARVC Registry.

Ongoing 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 will also be collected.

How to Join: Contact Crystal 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! Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu

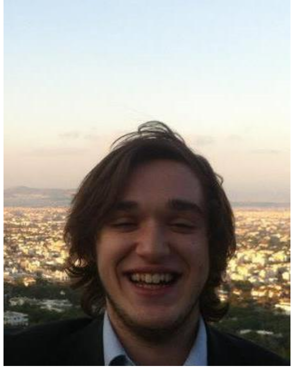
THANK YOU FOR YOUR PARTICIPATION IN ALL OF THESE IMPORTANT STUDIES!!!

Meet Our Research Interns

Anke Hodes

We had the pleasure of hosting Anke Hodes, a senior medical student from the University Medical Centre Groningen in the Netherlands. Anke joined us from September-January to do an honors research clerkship – the last step of her medical school training. Anke worked with the ARVD team and her Dutch advisor, prior ARVD/C seminar featured speaker J. Peter van Tintelen, MD, PhD, to investigate pregnancy outcomes in women with ARVD/C. Combining the pregnancy histories of Dutch and North American women allowed us to describe the outcomes of 45 pregnancies in women diagnosed with ARVD/C at the time of pregnancy. We found that most women and their babies do well – although several will have an arrhythmia during pregnancy. Anke is working on submitting a paper describing these findings for publication. We hope this work will help women with ARVD/C and their doctors evaluate whether pregnancy is right for them.

George Katritsis



George Katritsis, a senior British medical student at the University of Bristol (and originally from Greece) joined us for an 8-week research elective last summer. George focused his work on investigating the outcomes of three ARVD/C patients enrolled in our Registry who had undergone tricuspid valve repair or replacement for worsening heart failure symptoms. George is preparing a description of the outcomes of these surgeries for publication now. While the surgery did not halt ARVD/C progression, our patients felt better after the surgery. Tricuspid valve surgery may be an option for patients with severe right-sided heart failure.

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 would like to take this opportunity to make you aware of a blog started by the ARVD/C Youth Society. Several members of this group have shared their experiences which have been published in The Huffington Post. The ARVD/C Youth Society also host a private group on Facebook which is geared toward individuals with ARVD that are 30ish and younger. Their mission is to connect young ARVD patients with other ARVD patients around the world.

Read their stories here http://www.huffingtonpost.com/arvdc-youth-society/

Featured Manuscript

Impact of Genotype on Clinical Course in Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Associated Mutation Carriers

Aditya Bhonsale, MD; Judith A Groeneweg, MD; Cynthia James, PhD; Dennis Dooijes, PhD; Crystal Tichnell, MGC; Jan D Jongbloed, PhD; Brittney Murray, MS; Anneline S te Riele, MD; Maarten P van den Berg, MD, PhD; Hennie Bikker, PhD; Douwe E Atsma, MD, PhD; Natasja M de Groot, MD, PhD; Arjan C Houweling, MD, PhD; Jeroen F van der Heijden, MD, PhD; Stuart D Russell, MD; Pieter A Doevendans, MD, PhD; Toon A van Veen, PhD; Harikrishna Tandri, MD; Arthur A Wilde, MD, PhD; Daniel P Judge, MD; J Peter van Tintelen, MD, PhD; Hugh Calkins, MD; Richard Hauer, MD, PhD

Dr. Aditya Bhonsale is currently in the third year of his Cardiology Fellowship here at Johns Hopkins and has spent a significant amount of time studying the influence of genotype on the clinical course and arrhythmic outcome of patients with ARVD/C. This study evaluated the impact of pathogenic mutations in both desmosomal and non-desmosomal genes in 577 patients from the US and Dutch ARVD/C cohorts. We found that patients who presented with sudden cardiac death/ventricular fibrillation were younger than those patients who presented with sustained ventricular tachycardia. Patients with more than one mutation had earlier occurrence of arrhythmias and more significant disease. Patients who carried a mutation in DSP (Desmoplakin) were more likely to experience left ventricular disease and heart failure when compared to those patients who carried a mutation in PKP2 (Plakophilin-2). We followed 541 patients presenting alive over a period of 6 ± 7 years. During this time, 162 (30%) had sustained VT/VF episodes, 78 (14%) developed left ventricular dysfunction, 28 (5%) developed heart failure and 10 (2%) required cardiac transplant. Men were more likely to be the first person to present (proband) and with earlier and more severe arrhythmias.

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 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

THANK YOU!!!

A special thank you to everyone who has supported the Johns Hopkins ARVD/C Program over the past year. We truly appreciate the generosity of patients and families who have been able to support our program financially. We cannot thank you enough for your continued participation in responding to surveys, sending medical records, providing blood/skin samples, etc. Together, we will continue to solve the mysteries of ARVD/C.

Support Group Info

Looking for a support group?

ARVD support group on Yahoo:
https://groups.yahoo.com/neo/groups/mmettera/info
(Please note that this group will be migrating to Google Groups soon – search for "ARVD ARVC Support Group")
Any issues joining this group, email Bob at ralla52@yahoo.com

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** – private group

Coming soon…ARVD/C Mentor Program

Contact Us

Johns Hopkins Hospital
600 North Wolfe Street, Blalock 545
Baltimore, Maryland 21287
P: 410-502-7161, F: 410-502-9148
Website: www.**ARVD**.com