Johns Hopkins Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Program

Volume 7, Issue 2 Summer 2017

2017 ARVD/C Family Seminar

We had a beautiful weekend for this year's seminar and the most well-attended seminar to date with over 200 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. This was a great opportunity to interact with our staff and share stories with our attendees.

On Saturday, we started off with Brittney's introductory talk on The ABC's of ARVD/C. We then heard from a number of speakers addressing topics including diagnosis, management, genetics, disease mechanism, catheter ablation, sympathectomy, and psychosocial impact of ARVD/C on the patient and family.

A number of Hopkins faculty members presented including Dr. Hugh Calkins, Dr. Hari Tandri, Dr. Daniel Judge, Dr. Cynthia James. We were delighted to have Professor Firat Duru join us all the way from Switzerland. He shared his experience of ARVD/C in Switzerland, as well as the role of hormones. Dr. Ali J. Marian also joined us from The University of Texas to offer some insight into disease pathogenesis and treatment of ARVC from a basic science perspective. Last, but certainly not least, we were honored to have Dr. Samuel Sears, PhD from East Carolina University for the 7th time. He was accompanied by some of his students who helped lead discussion groups in the afternoon.

In addition, research opportunities were held throughout the afternoon, including blood draws, ICD interrogations, ECGs and Holters. Thank you to everyone who was able to stay and participate in the various research studies. You are a vital part of our research success.

View presentations now at:
http://tinyurl.com/2017ARVDSeminar

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JOHNS HOPKINS MEDICINE

Welcome New ARVD/C Program Team Members

Julia Cadrin Tourigny MD, has completed her medical education and training in Cardiology at the Université de Montréal and was certified by the Royal College of Physicians of Canada (FRCPC) in 2015. Following her strong interest in clinical research, she obtained a Master of Biomedical Sciences for her work on the efficacy of amiodarone in patients with atrial fibrillation and heart failure. She then pursued additional training in cardiac electrophysiology at the Montreal Heart Institute from 2015 to 2017. Over the past year, she has undertaken a postdoctoral clinical and research fellowship in inherited heart disease at the Johns Hopkins Hospital under the George Mines Traveling Fellowship from the Canadian Heart Rhythm Society. She has joined the ARVD/C group and leads an important multicentre project aiming to improve the arrhythmic risk stratification in patients with ARVC.

Weijia Wang, MD attended Zhejiang University in China for his medical education between 2005 to 2012. He finished his internal medicine residency at Tufts Medical Center in Boston and became board certified in 2016. He acquired a strong interest in clinical research and earned a Master of Public Health degree with a concentration in Epidemiology and Biostatistics from the Johns Hopkins Bloomberg School of Public Health in May 2017. Over the past year, Weijia worked closely with our group. The work on how exercise restriction affects disease outcomes in ARVD/C won the Clinical Research Award in Honor of Mark Josephson and Hein Wellens from the Heart Rhythm Society. He joined the ARVD/C group as a research fellow this July and will continue the work on understanding the impact of exercise in ARVD/C.

Anna Burton joined the ARVD/C team in June 2017 as the new Genetic Counselor Assistant. She is from Indiana and recently graduated from Denison University in Ohio. Her main roles include gathering our patients' medical records, inviting patients to join our research studies, and assisting the genetic counselors with other paperwork and administrative tasks.

Giovanna Jimenez joined the ARVD/C program in May 2017 as our Clinic Program Coordinator. While Giovanna is new to the ARVD/C team, she is not new to Hopkins. Prior to joining our team, Giovanna was a Senior Administrative Coordinator for the division, specifically to Dr. Hari Tandri. Giovanna is involved with scheduling our clinical patients so many of you will be interacting with her.

Dr. Nisha Gilotra, Assistant Professor of Medicine and Co-Director of the Heart Failure Bridge Clinic, is a cardiologist here at Hopkins with expertise in advance heart failure. She completed her medical residency, general cardiology fellowship and advance heart failure fellowship all here at Johns Hopkins. She has worked with ARVD/C Program research to assess the incidence of heart failure among our ARVD/C patients. This paper was just accepted for publication in Circulation: Heart Failure. Dr. Gilotra will now be primarily involved in assessing our patients on a clinical basis.

We had to say goodbye to our genetic counseling assistant, Bryana Rivers, in July as she was accepted into the Genetic Counseling Master's degree program at the University of Cincinnati. Best Wishes to Bryana!

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 specialist, Dr. Nisha Gilotra can also be arranged. These appointments are billed to your health insurance. To schedule an appointment, contact Crystal.

Tele – Genetic Counseling

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.

Pediatrics

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.

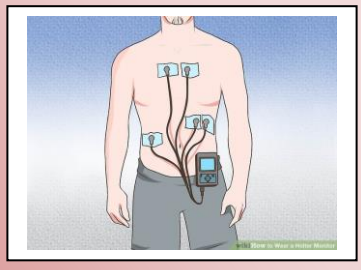
Crystal Tichnell, MGC ctichnell@jhmi.edu 410-502-7161

Clinical Updates – Holters and Metabolics

Why do I Need a Holter if I have an ICD?

Who can't wait to wear their yearly Holter monitor for 24 hours?? It's hot out, you can't sleep with it on, you can't get it wet, and you have this ICD that supposedly monitors every heartbeat, so why an extra monitor you ask?

The 24 hour Holter monitor is one of the most important screening tools we have to assess your current arrhythmia burden as well as to assess how well your current medication regime is in managing your arrhythmias. ICDs generally do not measure PVC (extra beats) burden, so this easy test is very important!



Metabolic Stress Tests

A metabolic stress test is a type of exercise test that measures how well your heart and lungs function during exercise. In most cases you exercise on a bike with a mouthpiece in and nose clips on. You are hooked up to an EKG monitor, as well as blood pressure throughout the test. The test starts off walking and gradually increases in intensity. It's important to go as long as you can to get an accurate assessment. This will help us in monitoring your function when assessing for signs and symptoms of heart failure.

We are starting to get baseline measurements on those patients that are followed here at Hopkins.

Ongoing Research Opportunities at Johns Hopkins

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 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! Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu.

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

Ongoing Research Opportunities at Johns Hopkins

Relative Safety, Efficacy, and Patient Satisfaction of Standard ICDs versus the Sub-Cutaneous ICD (S-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).
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

Once enrolled, please remember to check your email for reminders to complete the online questionnaires. It is really important that we continue to collect this follow up data. Thank you for your participation!

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)*

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Outcomes of genetic counseling for arrhythmogenic cardiomyopathy: A comparison of face-to-face and tele-genetic counseling

As we've mentioned before, we are now offering tele-genetic counseling! We are taking this opportunity to determine whether genetic counseling services offered via telephone/ videoconference differs from the traditional face-to-face genetic counseling in achieving three key cardiac genetic counseling outcomes: reducing cardiac-specific anxiety, increasing disease-specific genetic understanding, and enhancing patient empowerment. Data will be collected by completing questionnaires 2 weeks preceding and 2 weeks following a genetic counseling session for an arrhythmogenic cardiomyopathy indication. 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.

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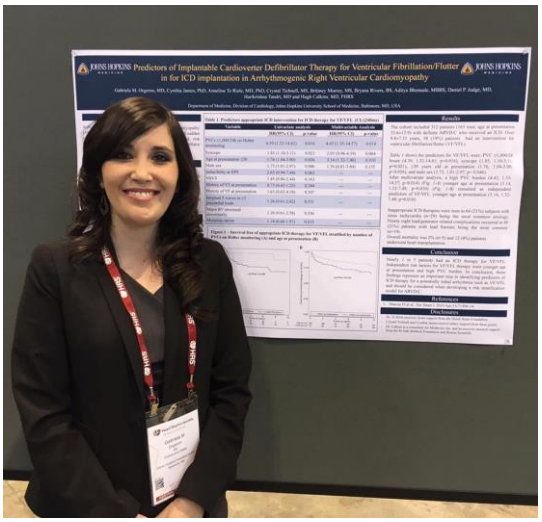
Featured Manuscript

IMPLANTABLE CARDIOVERTER-DEFIBRILLATOR THERAPY IN ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY: PREDICTORS OF APPROPRIATE THERAPY, OUTCOMES, AND COMPLICATIONS

Gabriela M. Orgeron, MD; Cynthia A. James, ScM, PhD; Anneline Te Riele, MD, PhD; Crystal Tichnell, MGC; Brittney Murray, MS; Aditya Bhonsale, MD; Ihab R. Kamel, MD, PhD; Stephan L. Zimmerman, MD; Daniel P. Judge, MD; Jane Crosson, MD; Harikrishna Tandri, MD; Hugh Calkins, MD. J Am Heart Assoc. 2017 Jun 6;6(6).



Gabriela M. Orgeron, MD

This study looked at 312 patients who met the task force criteria for definite ARVD/C and who also received an implantable cardioverter defibrillator (ICD). An important decision for someone diagnosed with ARVD/C is whether or not they should receive an ICD for the treatment of serious arrhythmias. This study looked at a number of factors to see if we could predict who might experience a serious arrhythmia. These included age at presentation, gender, mutation status, history of passing out, history of ventricular tachycardia (VT) at the time of presentation, history of nonsustained ventricular tachycardia or NSVT, inducibility at the time of an electrophysiology study (EPS), presence of inverted T-waves on ECG, more than 1000 PVCs on Holter monitor, and structural abnormalities fulfilling a major criteria.

The study also looked at rates of complications and inappropriate interventions. We also looked at long-term outcomes and survival.

The analysis showed that ARVD/C patients with ICDs have a remarkably high incidence of appropriate ICD therapy. The presence of T-wave inversions in more than 3 precordial leads, greater than 1000 PVCs on Holter, inducibility on EPS, history of sustained VT at presentation, and male gender were identified as predictors of an appropriate ICD therapy.

It is important to carefully identify those individuals at high risk of sudden cardiac death to ensure appropriate treatment. This also protects low-risk individuals from potential complications and unnecessary therapies. ICDs do save lives!

This full article is available for viewing at: https://doi.org/10.1161/JAHA.117.006242

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Recent Publications

Calkins H. ***The ventricular ectopic QRS interval for diagnosis and risk stratification in arrhythmogenic right ventricular dysplasia/cardiomyopathy: Is this the answer?*** Heart Rhythm. 2016 Jul;13(7):1513-4.

Rastegar N, Te Riele AS, James CA, Bhonsale A, Murray B, Tichnell C, Calkins H, Tandri H, Bluemke DA, Kamel IR, Zimmerman SL. ***Fibrofatty Changes: Incidence at Cardiac MR Imaging in Patients with Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy.*** Radiology. 2016 Aug;280(2):405-12.

Rhodes A, Rosman L, Cahill J, Ingles J, Murray B, Tichnell C, James CA, Sears SF. ***Minding the Genes: a Multidisciplinary Approach towards Genetic Assessment of Cardiovascular Disease.*** J Genet Couns. 2016 Sep 5. In Press.

Corrado D, Link MS, Calkins H. ***Arrhythmogenic right ventricular cardiomyopathy.*** N Engl J Med. 2017 Jan 5;376(1):61-72. Review.

Orgeron GM, Crosson JE. ***Arrhythmogenic right ventricular dysplasia/cardiomyopathy.*** Cardiol Young. 2017 Jan;27(S1):S57-S61.

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Mast TP, James CA, Calkins H, Teske AJ, Tichnell C, Murray B, Loh P, Russell SD, Velthuis BK, Judge DP, Dooijes D, Tedford RJ, van der Heijden JF, Tandri H, Hauer RN, Abraham TP, Doevendans PA, Te Riele AS, Cramer MJ. ***Evaluation of Structural Progression in Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy.*** JAMA Cardiol. 2017 Mar 1; 2(3):293-302.

Fiallos K, Applegate C, Mathews DJ, Bollinger J, Bergner AL, James CA. ***Choices for return of primary and secondary genomic research results of 790 members of families with Mendelian disease.*** Eur J Hum Genet. 2017 Mar 8.

Ingles J, James CA. ***Psychosocial care and cardiac counseling following sudden cardiac death in the young.*** 2017 Mar 1; In Press.

Haggerty CM, James CA, Calkins H, Tichnell C, Leader JB, Hartzel DN, Nevius CD, Pendergrass SA, Person TN, Schwartz M, Ritchie MD, Carey DJ, Ledbetter DH, Williams MS, Dewey FE, Lopez A, Penn J, Overton JD, Reid JG, Lebo M, Mason-Suares H, Austin-Tse C, Rehm HL, Delisle BP, Makowski DJ, Mehra VC, Murray MF, Fornwalt BK. ***Electronic Health Record Phenotype in Subjects with Genetic Variants Associated with Arrhythmogenic Right Ventricular Cardiomyopathy: A Study in 30,716 Subjects with Exome Sequencing.*** Genet Med. 2017 May 4.

Bhonsale A, Te Riele AS, Sawant AC, Groeneweg JA, James CA, Murray B, Tichnell C, Mast TP, van der Pols MJ, Cramer MJ, Dooijes D, van der Heijden JF, Tandri H, van Tintelen JP, Judge DP, Hauer RN, Calkins H. ***Cardiac phenotype and long-term prognosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia patients with late presentation.*** Heart Rhythm. 2017 Jun;14(6):883-891.

Orgeron GM, James CA, Te Riele A, Tichnell C, Murray B, Bhonsale A, Kamel IR, Zimmerman SL, Judge DP, Crosson J, Tandri H, Calkins H. **Implantable Cardioverter-Defibrillator Therapy in Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy: Predictors of Appropriate Therapy, Outcomes, and Complications**. J Am Heart Assoc. 2017 Jun 6;6(6).

Gupta R, Tichnell C, Murray B, Rizzo S, Te Riele AS, Tandri H, Judge DP, Thiene G, Basso C, Calkins H, James CA. ***Comparison of Features of Fatal versus Non-Fatal Cardiac Arrest in Patients with Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy.*** Am J Cardiol. 2017 Jul 1;120(1):111-117.

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

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Abigail Kemp

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February of 2016 was my first memorable arrhythmic encounter and the beginning of my journey to diagnosis.

It felt like my heart just sank –the room was starting to get dark and my ears started ringing. I thought to myself, "shit I'm going to pass out, this is so embarrassing". This was during a cycle class with a few of my friends. A few months later I road my bike to work and it happened again, then again during a jog around the park behind my house and a hike in the mountains. After that I started avoiding cardio all together. I wanted to pretend that nothing was wrong when in all reality those were giant warning signs.

Late spring of 2016 is when a new feeling started creeping into my life: anxiousness. Conceptually, I knew that there was no reason for me to be feeling these sensations in my chest but they were there and really started to take a psychological toll on my daily life. During this time period I had an IUD implanted and thought that anxiety was a possible side effect? During my follow-up appointment with my general practitioner, I asked her if that was a possibility. As our conversation continued I also mentioned my near fainting episodes. She was instantly alarmed by this and recommended I go see a cardiologist and get a full cardiac workup done. For me the near fainting was the least of my concerns, this feeling of anxiousness was what I was really concerned about.

Just before Labor Day weekend is when I went in for my first stress test, echo and 48 hour Holster monitor. I never thought at the age of 26 that I would be seeing a cardiologist and getting a stress test, people in their 50's do this. Again, my main concern was this anxious feeling that I was having. My stress test just showed that I was having PVC's and the doctor suggested that those could be causing me the feeling of anxiousness. The next 48 hours I was wired up and told to workout and push myself to see if they can catch anything on the monitor. After turning the monitor in and while waiting for the results, that feeling of anxiousness completely took me over again while I was at work. I immediately left work and drove myself home where my mom and I contemplated whether I should go to the ER. I now realize that this one contemplation could have been the difference between life and death for me. I did decide that day to go in and for the next 10 days I would call Porter Hospital "home."

I now have an ICD implanted to safeguard against the next time my heart decides to have another v-tach episode and I will be taking medication every day for the rest of my life to help keep the arrhythmias calm. My journey post-diagnosis is just beginning and has already been full of many ups and downs. What I have learned through this entire experience is to not let fear dictate the way I choose to life my life, I am a fighter with big ambitions, and most importantly I am so loved.

I cannot thank my general practitioner Dr. Theresa Holson, all of the staff at Porter Hospital, my cardiologist Dr. Choe and all staff at South Denver Cardiology, my psychologist Dr. Bloom, and my friends and family for their love and support.

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**, 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, 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

ARVC Program Staff

Hugh Calkins, MD—Director
Harikrishna Tandri, MD—Faculty
Cynthia James, ScM, PhD—Genetic Counselor
Brittney Murray, MS—Genetic Counselor
Crystal Tichnell, MGC—Genetic Counselor
Anna Burton—Genetic Counselor Assistant
Giovanna Jimenez—Clinic Coordinator
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 at ctichnell@jhmi.edu Thank you!

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CONTACT US

www.ARVD.com

Support Group Info

Looking for a support group?

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 for more information and visit ARVDHEARTANDSOUL.org

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