

JOHNS HOPKINS ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY PROGRAM

Volume 12, Issue 1

Summer 2022

* ** ***!!!Happy Summer!!!*** ** *

It's been just over two years since COVID-19 disrupted the world as we knew it. While we are slowly returning to some normalcy, it's important to not let your guard down. Continue to do your part to minimize the spread of COVID-19 to those who are most vulnerable.

Our annual ARVC patient and family seminar was held virtually again this year in March with nearly 500 registrants. This year we focused on *The Promise of Research in ARVC* where members of our research team presented the projects they have been working on and what the future may hold for ARVC treatment. The highlight of the seminar was learning about the promise of gene therapy in ARVC. Gene therapy is a technique that treats or prevents disease by correcting the underlying genetic alteration. Many companies are interested in learning more about ARVC caused by PKP2 pathogenic variants and apply gene therapy techniques as a potential therapy.

We had fantastic presentations by our research fellows on PVCs and Follow up - Why Holters Matter (Alessio Gasperetti); Heart Transplant in ARVC – A Growing but Unique Journey (Paul Scheel); Improving ARVC Diagnosis Through Machine Learning (Rick Carrick); and Desmoplakin (DSP), Myocarditis, and ARVC (Weijia Wang). In addition, genetics presentations on How Big-Data Population Genomics is Improving Our Understanding of the Genetics of ARVC (Cindy James) and Updates in ARVC Genetics (Brittney Murray) laid the groundwork for the promise of new therapies in ARVC. Shilpi Epstein described “Results from the First In-Human Trial of Gene Therapy Treatment for Danon Disease” and Whit Tingley presented on “Gene Therapy Approaches to ARVC”. We ended the program with Sam Sears sharing “3 Things that COVID Taught Everyone without ARVC”. Many of these presentations are available for viewing on our website, www.ARVD.com.

Attendees took advantage of self-moderated break-out sessions (Transplant Talk – Sharing Stories, Under 30 – Diagnosed with or at risk of developing ARVD, CareGivers and Support People, and Hope for ARVD/C – Face to Face Meeting) and various discussion boards, in an effort to network with others. Thank you to everyone who made this year's seminar amazing!

We continue to value the in-person conference setting and hope we are able to gather once again in Baltimore in Spring 2023. We've missed this so much and look forward to the in-person interaction again soon! Stay tuned!



~ The ARVD/C Program

Hugh Calkins, MD

Hari Tandri, MD

Cindy James, PhD

Brittney Murray, MS

Crystal Tichnell, MGC, RN

ARVD/C Clinical Services

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. All appointments are billed to your health insurance.

With the COVID-19 pandemic becoming less of an impact, licensure waivers are coming to an end. This means we are no longer able to offer telemedicine appointments as we were over the past year. However, there may be some flexibility with our genetic counseling ONLY visits to be able to continue to offer this option. Please reach out to Crystal to see if you are eligible for a telemedicine appointment based on your appointment needs and physical location. Our clinics are fully open so please reach out to get scheduled. Remember, even if your condition is stable, you should be checking in at least once every two years with repeat cardiac evaluations. It is best to respond to changes in your health, rather than react to an urgent situation.

To schedule an appointment, contact Crystal at ctichnell@jhmi.edu or 410-502-7161.



Research Opportunities at Johns Hopkins

The Importance of Clinical Trials

Clinical trials are so important in discovering new treatments and the safety of those treatments, particularly of rare conditions. The decision to participate in a clinical trial is a personal one and only you can decide if it's the right choice for you. As you think about whether to participate, please take a minute to consider the impact your participation or non-participation might have on overall clinical trials in ARVC. As we all know, ARVC is a rare condition and funding to support rare conditions is limited. We need to prove to big funding agencies that individuals with ARVC are interested and willing to participate in new discovery research, otherwise, there will be huge delays in new discoveries, new treatment options, and ultimately finding a cure. It is an exciting time for gene therapy and companies are considering ARVC. We will need your help to move this forward. Participation in our registry is the first, most important, step. Please make sure your information is up to date with our registry as this will determine your eligibility to participate.

<https://www.nih.gov/health-information/nih-clinical-research-trials-you/basics>

Clinical and Genetic Investigations of Right Ventricular Dysplasia (ARVD/C Registry)

This registry is the heart of our program and **from which all of our research projects originate**. This means eligibility for future clinical trials, including gene therapy, will require enrollment in our registry. You do not need to be a patient followed at Johns Hopkins to participate in our registry. Both children and adults either diagnosed with ARVC or a family member of someone diagnosed with ARVC are eligible to participate. Participation involves submission of past medical records and continued followup for at least 5 years (we will offer renewal for continued participation). A DNA sample may be collected for specific projects.

Reach out to Crystal at 410.502.7161 or ctichnell@jhmi.edu to join.

Featured Manuscript

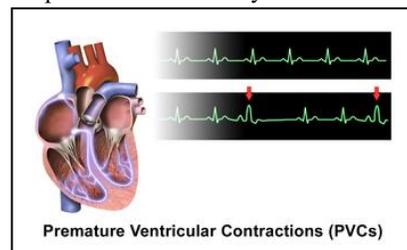
Association of Premature Ventricular Contraction Burden on Serial Holter Monitoring With Arrhythmogenic Risk in Patients With Arrhythmogenic Right Ventricular Cardiomyopathy

Gasperetti A, Cappelletto C, Carrick R, Targetti M, Tichnell C, Martino A, Murray B, Compagnucci P, Stolfo D, Bisson J, Gilotra N, Carbucicchio C, Olivotto I, Tandri H, Dello Russo A, Cadrin-Tourigny J, Calò L, Tondo C, Sinagra G, James CA, Casella M, Calkins H. JAMA Cardiol. 2022 Apr 1;7(4):378-385.

The Johns Hopkins ARVC program has been a strong proponent of Holters and ZioPatches for patient follow-up for years. Our experience was recently summarized in a study addressing this topic.

This study was a multicenter collaboration with ARVC centers in Canada and Italy, where 169 patients were followed up for a mean of 4.5 years and a grand total of 723 Holter/ZioPatches (on average 4/patients) was reviewed. Those numbers made study the single largest piece of scientific evidence addressing the use of multiple Holter in patients of ARVC currently available worldwide. The results of the study clearly showed how changes in the 24-hour burden of premature ventricular complexes (PVCs) at a Holter monitor could be used to dynamically track the individual risk profile of ARVC patients, even years after first diagnosis. In particular, sudden increases in the PVC burden at Holter/ZioPatch assessment ("PVC Spike") were associated with an increased risk of complex ventricular arrhythmias in the 12 months immediately following that evaluation.

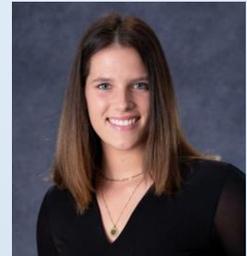
This study helped us proving how the performance of an Holter/ZioPatch monitor every 12-18 months is really important for the management of ARVC patients on the long run. Tracking PVCs over time helps clinicians constantly reassessing patient risk, providing them with data about the need for pharmacological titration of drugs, and complements many other tests (i.e. ECG and magnetic resonance) in the assessment of whether an ICD is needed in an individualized manner.



Genetic Counseling Student Projects

The Johns Hopkins ARVC Program is a primary clinical rotation site for students attending the University of Maryland and Johns Hopkins University/National Institutes of Health (JHU/NIH) genetic counseling training programs. In addition to rotations in pediatrics, prenatal, and cancer, students are exposed to specialty clinics, like cardiovascular genetics. Students attend a 4-6 week rotation where they learn the process of genetic counseling within the Center for Inherited Heart Disease, or specifically, the ARVC Program. You might have interacted with a genetic counseling student during one of your visits! As part of their training, students are also required to complete an independent study project. This year, we have 2 students working on projects related to ARVC. Their projects are summarized below.

Jessica Sweeney is a rising third year student at the Johns Hopkins/National Institutes of Health (JHU/NIH) Genetic Counseling Training Program. Her thesis project is entitled, "Characterizing Decision-Making Surrounding Exercise in Individuals with ARVC: analysis of decisional conflict, decisional regret, and shared decision-making. Jessica will be administering a survey to participants in the Johns Hopkins ARVC Registry that will retrospectively explore exercise decision-making at the time of diagnosis with ARVC or disclosure of at-risk status. The decision quality will be characterized by how well the participant feels that their decisional needs were met, and to what degree a shared decision-making process occurred. The main outcomes of the study will be decisional outcomes, as defined by the Ottawa Decisional Support Framework). These include what decision was made (i.e. the level of exercise participation), decisional regret, and decisional conflict. The proposed study will describe the exercise decision-making process for those diagnosed with or at-risk for developing ARVC, and inform genetic counselors on how to best support these patients through their decision-making process.



Emma Schopp is a rising second year genetic counseling student at the JHU/NIH Genetic Counseling Training Program. She is in the beginning stages of thinking about her thesis project and is interested in exploring perceptions of gene therapy treatment among individuals living with risk for cardiac events attributable to their genetics. Specifically, she is interested in learning how patients compare the uncertainty associated with living with their condition to the uncertainty of participating in a clinical trial for novel gene therapy. In addition, she is curious about other factors that may affect or moderate a patient's willingness to participate, such as distress about disease burden, specific disease experiences, anxiety, resources, support system, personality related factors, altruism, integration into the disease community, and trust in one's provider, the medical system, and science. Emma's plan is to organize some focus groups to see what patients are most interested in learning about the prospect of gene therapy.

We wanted to introduce these projects to you as we may be asking for your participation! **Thank you in advance!**



Hopkins ARVC at HRS22

The Heart Rhythm Society (HRS) hosted its annual meeting in San Francisco and is the largest gathering of heart rhythm professional from around the world. This was one of the first meetings held in-person since COVID-19 and we couldn't wait to see our friends and colleagues.

Several members of our team presented on ARVC and are listed below:

- **Hugh Calkins**, “Risk of Sudden Death in ARVC”
- **Cindy James**, “PKP2 Cardiomyopathy – Risk Prediction in ARVC”
“Predicting Who is Most at Risk of Sudden Cardiac Death”
“Alternative Models of Delivering Cardiac Genetic Counseling and Care”
- **Brittney Murray**, “How Do We Make Genetic Counseling Accessible”
“The Expanding Role of Allied Health Professionals in the World of Cardiogenetics”, moderator
- **Richard Carrick**, “Longitudinal Prediction of Ventricular Arrhythmias in Patients with Arrhythmogenic Right Ventricular Cardiomyopathy”
- **Alessio Gasperetti**, “Programmed Ventricular Stimulation as an Additional primary prevention risk stratification tool in arrhythmogenic right ventricular cardiomyopathy: a multinational study” and
“Discrimination of Arrhythmogenic Cardiomyopathy Phenotypes: A Path Towards Diagnostic Criteria Redefinition” (Research Fellowship Awardee)
- **Eric Carruth**, “Predicted Risk of Ventricular Arrhythmia in Individuals with desmosome gene variants identified via population genomic screening”
- **Stephen Chelko**, “Mechanisms of Immune-mediated injury in arrhythmogenic cardiomyopathy”. Stephen was also inducted as a Fellow of the Heart Rhythm Society or FHRS member.



Meet Our New Staff

We'd like to welcome several new staff members to the ARVC team. Zeba Shaik joined the ARVC Program in March as a research program coordinator. Zeba is originally from Boston, Massachusetts. She received her Master of Science in Physiology at Georgetown University in D.C. Her future plans include attending medical school and traveling to experience different foods, music, and cultures from all over the world. Zeba's primary role is working with our research database and collecting blood samples from those who join our research studies. You may see Zeba in clinic the next time you are here!



Leonore Okwara, MPH joined the team just this month as a research program manager. She will be managing the many tasks associated industry collaborations including contracts and budgets as they relate to clinical trials for new therapies in ARVC. Leonore has over 15 years of experience managing national research initiatives within the academic, corporate, and government sectors. She earned her B.A. in Sociology from Fort Hays State University and her Master of Public Health with a concentration in Epidemiology from Eastern Virginia Medical School. Leonore has extensive experience with the research lifecycle, grant development and management, budget management, and serving as a liaison between the community, researchers, and funders to identify priorities and achieve research goals. In her previous role, Leonore served as a Senior Program Manager for AllStripes, Inc. where she managed partnerships with rare disease Patient Advocacy Groups to engage their communities in research programs.



ARVD/C Program Info

ARVC Program Staff

Hugh Calkins, MD—Director
Harikrishna Tandri, MD—Co-Director
Nisha Gilotra, MD—Heart Failure
Caridad de la Uz, MD—Pediatrics
Stefan Zimmerman, MD—MR Imaging
Allison Hays, MD—Echo Imaging
Cynthia James, ScM, PhD—Genetic Counselor
Brittney Murray, MS—Genetic Counselor
Crystal Tichnell, MGC, RN—Genetic Counselor, Nurse
Emily Graham—Genetic Counselor Assistant
Christal Holmes-Igwebike—Clinic Coordinator
Catherine Pendelton—Research Program Coordinator
Alessio Gasperetti—Research Fellow
Zeba Shaik – Research Program Coordinator
Leonore Okwara – Research Program Manager

Keep us informed of your most up-to-date contact info!

Please send any changes and updated medical records to Crystal via the contact info below. Don't forget to schedule your routine follow up even if you feel fine.

Thank you!

Contact Us

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Website: www.ARVD.com
Email: ctichnell@jhmi.edu

How You Can Help

The Johns Hopkins ARVC/D Program provides world-class medical care. But **did you know** that the program relies heavily on outside donations to make a difference to ensure we are able to provide exceptional personalized care and to find more efficient and effective means to diagnose and treat our patients?



Did you know...FACT or MYTH?

- **We need money from philanthropy to support staff, research, patient care and education.**
FACT- We do! 75% of ARVC Program staff are financially supported by private funding/philanthropy.
- **The pandemic did not affect our program.**
MYTH- We continued consults throughout the pandemic thanks to telemedicine. Hospital wide; however, resources were diverted to caring for COVID-19 patients and elective procedures were significantly reduced. This overall reduced volume led to decreased revenues across the hospital and therefore reduced resources available to support the program.
- **It is “easy” to acquire grants/funding for ARVC research.**
MYTH- It is *much* more difficult to acquire outside funding through federal and private grants since ARVC is a rare disease. The Johns Hopkins Heart and Vascular Institute has the top ARVD/C Program in the country, under the leadership of Dr. Hugh Calkins and Co-Director, Dr. Hari Tandri. Patients and their families come to Johns Hopkins to receive an accurate diagnosis, treatment, genetic counseling, and family screening. We need **YOUR HELP** to continue this transformative work.
- **An ARVC diagnosis has a huge impact on overall mental health of patients and their families.**
FACT- We have plans to hire a mental health professional on our team to provide the support and resources that are so needed for this unique patient community. Studies have shown that patients with ARVC are at an elevated risk for anxiety. At least 30% of our patients have significant clinical anxiety and 10% are diagnosed with depression. We feel these numbers are underreported. This is long overdue and we need to do something NOW to support these mental health concerns.
- **We rely heavily on contributions from grateful patients, families and friends to help us seek new information, improve treatments, and ultimately, A CURE!**
FACT! Your gift truly makes a difference.
- **There are a variety of ways to donate and to support the program.**
FACT!
 - You can make an outright gift of cash or securities
 - Give in honor or in memory of a loved one
 - Give through IRA's, wills and trusts
 - Become a monthly donor
 - Be part of our online “Friend Raising” campaign. More info to follow!

To make a donation go to: <https://secure.jhu.edu/form/heart> (Select “**ARVD Research**” from the drop-down menu)

Or mail in donation to:

Johns Hopkins University and Medicine
Attn: Heart and Vascular Institute
PO Box 49143
Baltimore MD 21297-9143

(Be sure to include “ARVD Program” in the memo line of your check)

The Johns Hopkins Heart and Vascular Institute Development Office is here to help! Please contact Megan Lally, Associate Director of Development, at mlally2@jh.edu or 443-687-2947 for more information



Support Resources

Looking for support resources?

FACEBOOK Groups (private):

- ARVD/C Youth Society
- Hope for ARVD

PLN Heart Disease Foundation:

<https://www.plnheartdiseasefoundation.org>

Information for those of you affected by PLN or phospholamban type ARVC.

Precision Medicine Website:

Read "Feeling the Beat": Stories from the ARVC Community here:

<http://www.hopkinsmedicine.org/inhealth/precision-medicine-centers/arvc>

Sudden Arrhythmia Death Syndromes Foundation (SADS):

www.sads.org

An advocacy group for patients and families affected by a sudden arrhythmia death syndromes, including ARVC.



Sudden Arrhythmia Death Syndromes (SADS) Foundation

We continue to partner with the SADS Foundation to provide resources to our families impacted by ARVC. Please consider taking advantage of some of the support resources SADS has to offer, including ICD support groups and Living with SADS webinar series. Even if you could not attend these webinars live, I highly encourage you to go back and watch the recordings. Most importantly, their annual conference is coming up in July and will be held virtually for all to attend. Visit www.sads.org for more information on these excellent resources.



If you have a topic or information you'd like to see discussed in the next newsletter, please reach out to Crystal Tichnell at ctichnell@jhmi.edu

Thanks for reading!