Johns Hopkins Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Program

Volume 11, Issue 2 **Summer 2021**

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

COVID-19 has presented us with many challenges over the last year, but we have persevered and finally see the light at the end of the tunnel. I encourage you to continue to find the positive influence this pandemic has had on you personally and as a community. We can get so side-tracked with the negativity and that's not where we want or need to be.

Here at Johns Hopkins, the ARVC program continues to be hard at work with both local and international collaborators. We've learned to love Zoom so we can continue those important relationships. As maintaining your regular cardiac screenings is essential in a chronic condition, we were excited to be able to offer telemedicine options where permitted by state law. We are now seeing patients mostly in-person which has been refreshing. We've missed you!

Our most exciting accomplishment during the pandemic was the annual ARVC patient and family seminar. While we were unable to gather in Baltimore, we were able to host a virtual conference with the help of 14 ARVC international expert speakers, including: Dominic Abrams, Shannon Hourigan, Peter van Tintelen, Sam Sears, Cristina Basso, Mario Delmar, Jeff Saffitz, Corinna Brunckhorst, Kristina Haugaa, Julia Cadrin-Tourigny, Perry Elliott, Chris Semsarian, Jodie Ingles, and Andre LaGerche. The virtual format allowed us to reach 700 attendees from around the world! Attendees took advantage of break-out sessions (Phospholamban (*PLN*) focused discussion group with Peter van Tintelen and Dean Jensen, Heart Transplant Stories from patients, and a group for those diagnosed with ARVC under the age of 30) and various discussion groups from exercise to transplant, in an effort to network with others. We had fantastic presentations on Genetics and Genetic Testing, Pregnancy, How to Talk to Children and Teens, Seeking Your Best Possible Self, Progression, ICDs, and Exercise. Thank you to everyone who made this year's seminar amazing! Some of these presentations will also be posted on our website in the coming weeks www.ARVD.com.

We continue to value the in-person conference setting and hope we are able to gather once again in Baltimore in Spring 2021. However, the participation of both international patients and families, as well as ARVC experts and collaborators, was so phenomenal, that we will be considering a virtual format every few years. Stay tuned!

~ The ARVD/C Program

Hugh Calkins, MD

Hari Tandri, MD

Cindy James, PhD

Brittney Murray, MS

Crystal Tichnell, MGC, RN

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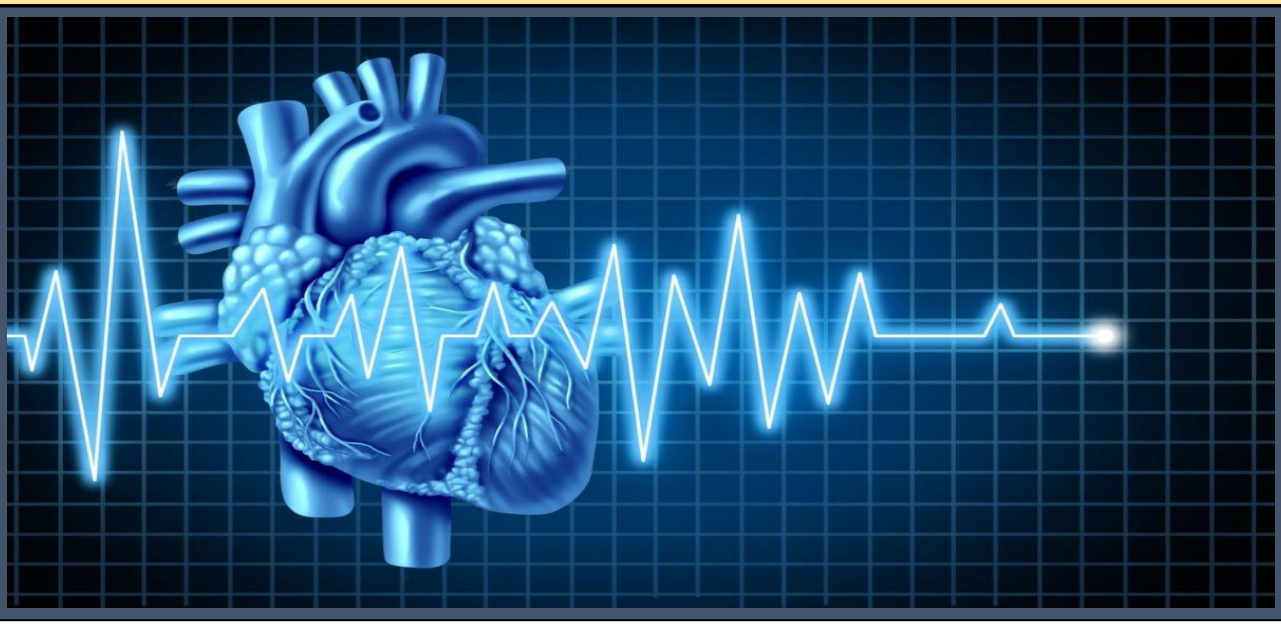
ARVD/C Clinical Services

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. These 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.

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



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Effects of Flecainide on Cardiac Arrhythmias in ARVC Patients

Johns Hopkins IRB00197430

Funded by The National Institute of Health (NIH)
Principal Investigator: Hugh Calkins, MD
Enrolling Site Coordinator: Crystal Tichnell, MGC, RN

The purpose of this study is to assess the effect of the antiarrhythmic drug, flecainide, on cardiac arrhythmias in individuals with ARVC. Participation in this study will last for about 10 weeks. This is a randomized, double-blinded, crossover study which means for part of the study you will be given the study drug, flecainide, and for the other part of the study you will be given a placebo. After 28 days you will switch, so everyone will be given the study drug at some point in the study. Neither you nor your enrolling physician will know if you are given the study drug or placebo first and that process is randomized.

During the study you will be asked to wear a monitor for 7 days on 2 occasions. We will also obtain ECGs and blood draws to assess flecainide levels through a home visit. Remote device interrogations will be obtained by your enrolling center. This study requires **one** in-person visit to discuss your enrollment and to sign the consent form.

To participate in this study you must:

- Be 18 years of age
- Meet the diagnostic criteria for ARVC
- Have a minimum of 500 PVCs on a recent 24-hour Holter monitor
- Have an implantable cardioverter defibrillator (ICD) with remote interrogation capability
- Be on a beta-blocker such as metoprolol, propranolol, atenolol, nadolol, carvedilol, unless contraindicated
- If prescribed, be willing to discontinue sotalol, quinidine, procainamide, propafenone, disopyramide, dronedarone, phenytoin, or mexilitene for 5 days with subsequent repeat 24 hour Holter
- Agree not to use any another antiarrhythmic medication during the 10 weeks of participation, unless for the management of life-threatening arrhythmias
- Agree to use medically acceptable contraceptive measures during participation unless documented as surgically sterile or post-menopausal

If you would like to learn more about this study or to discuss your eligibility, please contact Crystal Tichnell, MGC, RN at ctichnell@jhmi.edu or 410-502-7161. This is an important first clinical trial in ARVC and will pave the way for future clinical drug trials.

Research Opportunities at Johns Hopkins

Why Participate in a Clinical Trial?

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 there isn't much funding for research. So, whatever funding we do get, we need to make the most of it. 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.
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. 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. A DNA sample may be collected for specific projects. Reach out to Crystal at 410.502.7161 or ctichnell@jhmi.edu to join.

Featured Manuscripts

Clinical outcomes of catheter ablation of ventricular tachycardia in patients with arrhythmogenic right ventricular cardiomyopathy: Insights from the Johns Hopkins ARVC Program

Usama A. Daimee, MD, Fabrizio R. Assis, MD, Brittney Murray, MS, CGC, Crystal Tichnell, MGC, RN, Cynthia A. James, ScM, PhD, Hugh Calkins, MD, FHRS, Harikrishna Tandri, MD

The purpose of this study was to provide new insights on clinical outcomes based on a large series of VT ablation procedures from the current era in ARVC patients. Evaluating consecutive patients with definite ARVC who underwent ablation procedures between 2009 and 2019 at Johns Hopkins showed that after a single ablation procedure 68.6% and 49.8% of patients did not have VT after 1 and 5 years, respectively. Cumulative VT-free survival after multiple procedures was 81.8% and 69.6% at 1 and 5 years, respectively. In addition, the use of anti-arrhythmic medications decreased after ablation.

Arrhythmogenic Right Ventricular Cardiomyopathy Presenting as Clinical Myocarditis in Women

Paul J. Scheel, III, MD, Brittney Murray, MS, Crystal Tichnell, MGC, RN, Cynthia A. James, PhD, Harikrishna Tandri, MBBS, MD, Hugh Calkins, MD, Stephen P. Chelko, PhD, and Nisha A. Gilotra, MD

The focus of this study was to describe clinical myocarditis as an initial ARVC presentation and discuss the distinct clinical and genetic characteristics associated with this type of presentation. This cohort consisted of 12 patients (all female) referred between 2014 and 2019 diagnosed with myocarditis at presentation who were subsequently diagnosed with ARVC by Task Force Criteria. Majority presented with chest pain or ventricular arrhythmias. All patients had troponin elevations with reduced left ventricular function in 5 individuals. MRI demonstrated delayed gadolinium enhancement in the left ventricle and/or pericardial enhancement in 10 patients. Pathogenic variants were identified in 11 (10 in desmoplakin (*DSP*) and 1 desmoglein-2 (*DSG2*)). This study highlighted the important role of genetic testing among those with a myocarditis presentation to ensure an accurate clinical diagnosis.

SADS Partnership

We continue to partner with the Sudden Arrhythmia Death Syndrome (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 some ICD support groups and upcoming webinars. Visit www.sads.org for more information.

ICD Support Group

This virtual support group is a safe, open space to talk about your experience living with an ICD and meet your peers who know just what you're going through! This support group is facilitated by patients for patients. No medical questions will be answered.
Sign up for our ICD Support Group:
https://www.sads.org/living-with-sads/Support-Groups/ICD-Support-Group#.YMNnob6SnIU

Youth ICD Support Group

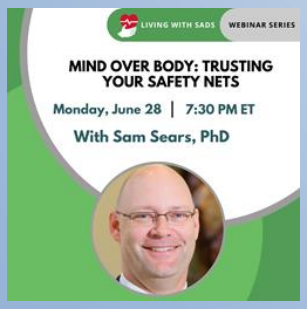
This virtual support group for kids or teens is a great way to connect with others around your age group who are also living with a SADS condition and an ICD. It is designed to provide social interaction with others who have a similar diagnosis. It will include fun, age-appropriate games and activities. Kids group for ages 9-13; Teen group for ages 14-18.
Sign up for our Youth ICD Support Group:
https://www.sads.org/living-with-sads/Support-Groups/Youth-ICD-Support-Group#.YMNnz76SnIU

Emotional support is crucial when living with a cardiac arrhythmia and /or the sudden loss of a loved one due to a cardiac arrhythmia. We encourage you to join and/or organize support groups that will facilitate emotional support, learning, healing, and increased knowledge. Because SADS diseases are not common and members are geographically spread out, online support groups are often the easiest way to connect with others who also have SADS diseases. Consider joining the SADS Foundation Facebook page as well.

Living with SADS – A Webinar Series

Register Here: https://www.sads.org/living-with-sads/Family-Webinars#.YMNobL6SnIU

Hear from medical experts, Dr. Harikrishna Tandri, Johns Hopkins ARVC program co-director, and Dr. Michael Ackerman, genetic cardiologist, Mayo Clinic about ablation and cardiac sympathetic denervation therapy on June 15 at 7:30 PM ET.



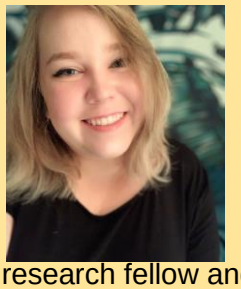
FREE WEBINAR SERIES

Dr. Sam Sears, a cardiac psychologist of East Carolina University, has spent much of his career working with heart patients with ICDs to overcome their anxieties. Join our webinar on June 28 at 7:30 PM ET to hear tips on how to improve your mental health and live and thrive with your cardiac condition.

Meet Our New Staff

We'd like to welcome several new staff members to the ARVC team. Congratulations to Emily Krupa, our current genetic counseling assistant, who will be joining University of Maryland's Genetic Counseling Program this Fall to start her career in genetic counseling.

This summer we will welcome Emily Graham to take over the genetic counseling assistant position. She is a 2019 graduate of University of Tennessee with a BA in Neuroscience and minor in Biology and Psychology. She has spent the past 2 years working as a lab tech in the University of Maryland Pharmacology lab. She is passionate about genetics outreach in underserved areas.



We are also excited to welcome Catherine Pendleton, our new research program coordinator, also joining us this summer. Catherine graduated from Dickinson College in 2018 with a BS in Biology. She is interested in exploring career options in the genetic counseling field and has come to the right place! She will be working on maintaining our IRB regulatory information and data abstraction, among many other research needs.

Alessio Gasperetti is our current ARVC post-doctoral research fellow and joined the ARVC Program in the Fall of 2020 from Italy. His current focus was recently funded by the Heart Rhythm Society to collect the largest cohort of patients with phenocopies of ARVC (specifically sarcoidosis, myocarditis, and phospholamban) and to compare their clinical characteristics with patients diagnosed with both classical (gene-elusive and *PKP-2* ARVC) and non-classical forms of ARVC. This will be another collaborative effort with Utrecht, Zurich, and Italy. The goal is to better refine the diagnostic criteria to tease out phenocopies of ARVC and to be more inclusive of arrhythmogenic cardiomyopathies with a more predominant left-sided characterization.

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

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.
Thank you!

Contact Us

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Baltimore, Maryland 21287
P: 410-502-7161, F: 443-873-5073
Website: www.**ARVD**.com
Email: ctichnell@jhmi.edu

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How You Can Help

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** 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 or arose25@jhmi.edu, for more information.

Ways to Make a Gift:

Online contributions:

https://secure.jhu.edu/form/heart (*Select "ARVD Research" from the drop-down menu)*

Contributions by Mail during the pandemic:

Checks can be made out to "**Johns Hopkins Medicine**" and sent to the address below.
*Please indicate "**ARVD Program**" as the designation of your gift on the memo line.*

Johns Hopkins at Keswick
Office of Advancement Services
3910 Keswick Road, Suite N2100
Baltimore, MD 21211
Attn: RevMgmt-HVI

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Support Resources

Looking for support resources?

FACEBOOK Groups (private):
- ARVD/C Youth Society
- Hope for ARVD

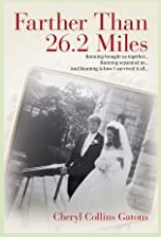
PLN Heart Disease Foundation:
https://www.plnheartdiseasefoundation.org
Information for those of you affected by PLN or phospholamban type ARVC.

Precision Medicine Website:
Read "Feeling the Beat": Stories from the ARVC Community here:
http://www.hopkinsmedicine.org/inhealth/precision-medicine-centers/arvc

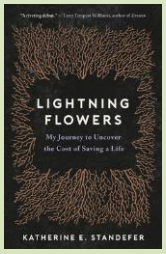
Sudden Arrhythmia Death Syndromes Foundation (SADS):
www.sads.org
An advocacy group for patients and families affected by a sudden arrhythmia death syndromes, including ARVC.

Published Personal Stories:

Racing Heart: A Runner's Journey of Love, Loss and Perseverance
By Kate Mihevc Edwards



Farther Than 26.2 Miles
By Cheryl Collins Gatons
can be purchased from Amazon



Lightning Flowers: My Journal to Uncover the Cost of Saving a Life
By Katherine E. Standefer

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!

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