Johns Hopkins Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Program

Volume 11, Issue 1 **Winter 2021**

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

A new year is upon us! What vision do you have for 2020? Well, these were the first 2 sentences to start off last year's newsletter. What was your vision for 2020? I bet you never imagined a pandemic shutting down our world. COVID-19 is what 2020 revolved around. Approaching almost a year of navigating the mysteries behind this virus, we are now starting to vaccinate and hoping to see an end to this pandemic.

Many routine activities have been altered by COVID-19 and for many, things slowed down a bit. I hope you were able to spend some time with family, start new hobbies, and find new ways of navigating life in the midst of a pandemic. For the ARVC program, our work never stopped. While enrollment for clinical trials may have slowed due to the challenges of a virtual environment, data abstraction and analysis remains on track and new collaborations continue to develop. From a clinical standpoint, I think we've probably seen even more patients with the telemedicine waivers in place for many states across the country. I hope many of you have been able to take advantage of telemedicine where state laws allow it.

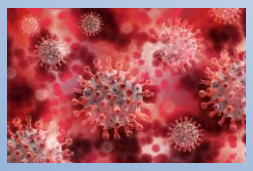
While our 2020 ARVC seminar was canceled due to COVID-19 and gathering restrictions, we were able to present a virtual mini-session in joint collaboration with the SADS Foundation. I hope many of you were able to attend. With the continued uncertainty about where we will be in Spring with regard to COVID-19, we are planning a virtual 2021 Patient and Family ARVC Conference on May 1st, 2021. It will certainly be difficult to not see you all in person for 2 years in a row, but for the safety of everyone, we feel it is the right decision at this time. With that being said, we do have a real treat for you this year. Given the virtual nature, we have invited over 15 international experts on ARVC and friends of the Johns Hopkins ARVC program to share short presentations on how they are helping to move the field of ARVC forward.

Registration for the seminar will be conducted through the WHOVA platform that we will be using for the seminar. To view the presentations, you will need to be registered for the event whether you are able to view during the live presentation on May 1st, 2021 or plan to view later. Registration details are included in this newsletter.

From all of us in the ARVC Program, we wish you good health, happiness, and success in the coming year and always!

COVID-19 Vaccine

Efforts have begun to distribute the COVID-19 vaccine. Please understand that this varies by state and based on available vaccine doses. When the time comes and you are offered a COVID-19 vaccine, we are recommending that you consider getting vaccinated. Based on our current knowledge, there appears to be no contraindication to getting the vaccine based on a diagnosis of ARVC. Should you require documentation certifying your diagnosis to be considered as someone with a high-risk medication condition, please **send your written request to Crystal at ctichnell@jhmi.edu.** We are happy to provide current Johns Hopkins patients diagnosed with ARVC that have been seen within the past 2 years with a letter certifying their medical condition. If you do not fall into this category (ie., you carry a genetic variant putting you at increased risk of developing ARVC but do not have evidence of disease OR have not had a clinical appointment with a member of our team within the past 2 years) you will need to discuss this request for documentation with your local providers. Please do not request this letter until you are offered an opportunity to receive the vaccine per your state guidelines. To start, each state is prioritizing its own residents. You will still need to continue precautions such as mask wearing, hand washing, social distancing, even once you are vaccinated based on current knowledge.



JOHNS HOPKINS MEDICINE

22nd Annual ARVD/C Patient and Family Seminar

Presented by
The Johns Hopkins ARVD/C Program

Due to the ongoing COVID-19 pandemic, this year's seminar will be held virtually on Saturday, May 1st, 2021. While we are incredibly disappointed to miss another year of gathering together in Baltimore, we are taking advantage of the virtual nature and inviting a large number of ARVC experts from all around the world. We will present both pre-recorded talks, as well as some live Q & A sessions. As we will not be presenting our standard basic talks, we encourage you to take some time to preview recordings from our most recent mini-session in collaboration with the SADS foundation. I highly recommend watching Brittney's ABCs of ARVD/C talk prior to the conference on May 1st. You can find this and the other presentations from the November Mini-Session on our website, www.arvd.com, under News and Events.

All communication regarding the seminar will be through the WHOVA online platform. Be sure to register and download the App so that you can take advantage of networking and viewing the agenda. Take a look below at the invited faculty.


Dominic Abrams, MD
Boston Children's Hospital


Cristina Basso, MD, PhD
University of Padua


Connie Bezzina, PhD
University of Amsterdam


Corinna Brunchkhorst, MD
Univ Hospital Zurich


Julia Cadrin-Tourigny, MD
Montreal Heart Institute


Mario Delmar, MD
NYU Langone Health


Perry Elliott, MD
Univ College London


Kristina Haugaa, MD
Oslo Univ Hospital


Shannon Hourigan, PhD
Boston Children's Hospital


Jodie Ingles, PhD
Centenary Institute


Andre La Gerche, MBBS, PhD
Baker Heart & Diabetes Institute


Sylvia Priori, MD, PhD
University of Pavia


Jeff Saffitz, MD, PhD
Beth Israel Deaconess Med Ctr


Samuel Sears, PhD
East Carolina University


Chris Semsarian, MBBS, PhD
Centenary Institute


Peter van Tintelen,MD, PhD
Univ Med Ctr Utrecht

Registration will go live on February 1st using this link:
ARVC Patient and Family Seminar Registration | Whova
https://whova.com/portal/registration/apafs_202105/

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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 Manuscript

Sudden Cardiac Death Prediction in Arrhythmogenic Right Ventricular Cardiomyopathy: A Multinational Collaboration Cadrin-Tourigny J et al

A few years ago we joined with colleagues from around the world to develop a statistical model to predict the chance that a patient newly diagnosed with ARVC who had never had a sustained ventricular arrhythmia (VA) - would develop a first VA in the first 5 years after diagnosis.

Now we have extended this study to develop a second statistical model that can be used in all definite newly diagnosed ARVC patients to predict the chance a patient will specifically have a fast ventricular arrhythmia. This is important because these fast VAs (which we are calling LTVA – for "life-threatening ventricular arrhythmia) are the most likely to cause sudden cardiac arrest.

We included data from 864 definite ARVC patients from 15 centers in North America and Europe. Association of 8 pre-specified clinical predictors with LTVA were tested. These included age, sex, prior sustained VA at any rate, recent history of fainting, 24-hour premature ventricular complexes count, the number of anterior and inferior leads with T-wave inversion on ECG, and both left and right ventricular ejection fraction.

Over an average 5.75 years of follow-up, 93 (10.8%) patients experienced LTVA. Of the 8 prespecified predictors, only 4 (younger age, male sex, premature ventricular complex count, and number of leads with T-wave inversion on ECG) predicted likelihood of LTVA. Importantly, prior sustained VA did not predict subsequent LTVA. We concluded that LTVA events in patients with definite ARVC can be predicted by a novel simple prediction model using only 4 clinical predictors.

Please check out our updated "risk calculator" at arvcrisk.com that includes both the first VA and LTVA risk predictions.

2020 Publications

Cadrin-Tourigny J, Bosman LP, Wang W, Tadros R, Bhonsale A, Bourfiss M, Lie ØH, Saguner AM, Svensson A, Andorin A, Tichnell C, Murray B, Zeppenfeld K, van den Berg MP, Asselbergs FW, Wilde AAM, Krahn AD, Talajic M, Rivard L, Chelko S, Zimmerman SL, Kamel IR, Crosson JE, Judge DP, Yap SC, Van der Heijden JF, Tandri H, Jongbloed JDH, van Tintelen JP, Platonov PG, Duru F, Haugaa KH, Khairy P, Hauer RNW, Calkins H, Te Riele ASJM, James CA. **Sudden Cardiac Death Prediction in Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC): A Multinational Collaboration.** Circ Arrhythm Electrophysiol. 2020 Dec 9.

Agrimi J, Scalco A, Agafonova J, Williams Iii L, Pansari N, Keceli G, Jun S, Wang N, Mastorci F, Tichnell C, Murray B, James CA, Calkins H, Zaglia T, Paolocci N, Chelko SP. **Psychosocial Stress Hastens Disease Progression and Sudden Death in Mice with Arrhythmogenic Cardiomyopathy**. J Clin Med. 2020 Nov 24;9(12):3804.

Costa S, Medeiros-Domingo A, Gasperetti A, Akdis D, Berger W, James CA, Ruschitzka F, Brunckhorst C, Duru F, Saguner AM.**Impact of Genetic Variant Reassessment on the Diagnosis of Arrhythmogenic Right Ventricular Cardiomyopathy Based on the 2010 Task Force Criteria**. Circ Genom Precis Med. 2020 Nov 24.

Gasperetti A, James CA, Cerrone M, Delmar M, Calkins H, Duru F. **Arrhythmogenic right ventricular cardiomyopathy and sports activity: from molecular pathways in diseased hearts to new insights into the athletic heart mimicry**. Eur Heart J. 2020 Nov 17:ehaa821.

Assis FR, Tandri H.**Epicardial Ablation of Ventricular Tachycardia in Arrhythmogenic Right Ventricular Cardiomyopathy**. Assis FR, Tandri H. Card Electrophysiol Clin. 2020 Sep;12(3):329-343.

Wang W, Tichnell C, Murray BA, Agafonova J, Cadrin-Tourigny J, Chelko S, Tandri H, Calkins H, James CA. **Exercise restriction is protective for genotype-positive family members of arrhythmogenic right ventricular cardiomyopathy patients**. Europace. 2020 Aug 1;22(8):1270-1278.

Bluemke DA, James CA, Te Riele ASJM. **Risk Stratification in Arrhythmogenic Right Ventricular Cardiomyopathy: Not a One-Size-Fits-All Strategy**. J Am Coll Cardiol. 2020 Jun 9;75(22):2766-2768.

Bosman LP, Cadrin-Tourigny J, Bourfiss M, Aliyari Ghasabeh M, Sharma A, Tichnell C, Roudijk RW, Murray B, Tandri H, Khairy P, Kamel IR, Zimmerman SL, Reitsma JB, Asselbergs FW, van Tintelen JP, van der Heijden JF, Hauer RNW, Calkins H, James CA, Te Riele ASJM.**Diagnosing arrhythmogenic right ventricular cardiomyopathy by 2010 Task Force Criteria: clinical performance and simplified practical implementation.** Europace. 2020 May 1;22(5):787-796.

Malik N, Win S, James CA, Kutty S, Mukherjee M, Gilotra NA, Tichnell C, Murray B, Agafonova J, Tandri H, Calkins H, Hays AG.**Right Ventricular Strain Predicts Structural Disease Progression in Patients With Arrhythmogenic Right Ventricular Cardiomyopathy**. J Am Heart Assoc. 2020 Apr 7;9(7):e015016.

Scheel PJ 3rd, Florido R, Hsu S, Murray B, Tichnell C, James CA, Agafonova J, Tandri H, Judge DP, Russell SD, Tedford RJ, Calkins H, Gilotra NA. **Safety and Utility of Cardiopulmonary Exercise Testing in Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia**. J Am Heart Assoc. 2020 Feb 4;9(3):e013695.

Brun F, Gigli M, Graw SL, Judge DP, Merlo M, Murray B, Calkins H, Sinagra G, Taylor MR, Mestroni L, James CA. **FLNC truncations cause arrhythmogenic right ventricular cardiomyopathy**. J Med Genet. 2020 Apr;57(4):254-257.

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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.

Due to the current COVID-19 pandemic, we are also offering telemedicine appointments with our team. Please note that the telemedicine laws and current licensure waivers due to COVID, vary by state. Please reach out to Crystal to see if you are eligible for a telemedicine appointment based on your appointment needs and physical location.

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

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 Krupa—Genetic Counselor Assistant

Christal Holmes-Igwebike—Clinic Coordinator

Rohan Mangal—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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Looking for support resources?

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

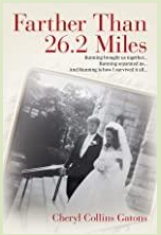
PLN Heart Disease Foundation:
https://www.plnheartdiseasefoundation.org
Information for those of you affected by PLN or phospholamban type ARVC.

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

Published Personal Stories:

Racing Heart: A Runner's Journey of Love, Loss and Perseverance
by Kate Mihevc Edwards *can be purchased from Amazon or through katemihevcedwards.com*

Kate was a passionate runner for years and lived for endurance sports. Until the day doctors told her she'd have to stop—or possibly die. After being diagnosed with ARVC, Kate was forced to mourn the loss of a lifestyle she loved and face a very scary question: What now? With the grit and determination that made her such an accomplished runner, Kate embarked on a journey that taught her to let go of her former self and claim a new life filled with strength, gratitude, and peace. ***Racing Heart*** *is Kate's testament to this journey as she walks readers through her transition from a runner eager to finish the race to a woman who values the preciousness of the present. Written for anyone facing a major life change or hungering for a life beyond the mundane,* ***Racing Heart*** *is the inspirational reminder that it is often from the most painful experiences that true joy and passion emerge.*



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

This is a true story of Cheryl, who went from running simply for the fun of it to qualifying for the Olympic marathon trials, thrusting her into the world of competitive running. It was in that world that Cheryl met another competitive runner, Kevin, and the two fell in love. Their shared passion for running brought the pair together, but it also tore them apart, as Kevin passed away (later determined to be from ARVD) navigating the state cross-country course with an athlete he coached. Cheryl shares her difficult journey in ***Farther Than 26.2 Miles****—how running brought her to the love of her life, how it took him away from her, and how it helped her survive after he was gone.*