

THE JOHNS HOPKINS ARRHYTHMOGENIC CARDIOMYOPATHY (ARVC/ALVC/ACM) PRECISION MEDICINE CENTER OF EXCELLENCE

Summer 2026

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WHAT'S INSIDE:

- 2026 Seminar Update
- Clinical Services at Johns Hopkins
- Heart Rhythm Society 2026
- SADS Foundation Family Conference
- Research Opportunities
- Featured Manuscripts
- ARVC Program Information
- Support the ARVC Program



OUR GOALS

Established in 1999, the ARVC program has 3 main goals:

- **EDUCATION OF PATIENTS AND PHYSICIANS** (newsletter, annual conference, arvd.com, publications, presenting at national conferences.)
- **FACILITATE THE EVALUATION AND MANAGEMENT OF PATIENTS WITH KNOWN OR SUSPECTED ARVC** (cardiac evaluations, genetic testing, genetic counselor / electrophysiologist / heart failure consultations)
- **PROVIDE NEW KNOWLEDGE ABOUT ARVC** (collaborative research and publications)

SEMINAR RECAP

Happy Summer from the Johns Hopkins ARVC/ACM Program! We hope you are able to enjoy some rest and relaxation this summer surrounded from friends and family.

We hosted our annual ARVC Patient and Family Seminar on April 17th and 18th, 2026. This year, we focused on offering an in-person only option. We truly value the opportunity for families to meet in community and develop friendships with others who have gone through similar experiences. We were able to record the seminar, so those who could not attend in-person will still have the opportunity to hear the information presented. We had nearly 90 attend the meet 'n greet Friday evening. Thank you to the Shah Family for sponsoring the event for the next 5 years! About 170 attended the conference on Saturday. Our speakers presented on all topics including ARVC diagnosis, genetics, and management, etc., with featured presentations by Sam Sears and Adam Helms. In addition, several industry representatives were in attendance to answer questions about their gene therapy focused trials. We continue to engage in conversations and collaborative efforts with industry so they can be better informed about the natural history of ARVC and how best to design future therapies. It is important for them to hear the patient perspective. The Sudden Arrhythmia Death Syndrome (SADS) Foundation was also present and continues to be an important support resource for patients.

The agenda was packed with presentations by the Johns Hopkins staff: Hugh Calkins, MD (Welcome and Update on the Johns Hopkins ARVC Program); Brittney Murray, MS (Back to Basics and Living Life with ARVC); Adam Helms, MD (Developing Treatments for the Root Causes of Arrhythmogenic Cardiomyopathy); Lili Barouch, MD (Strength vs Endurance Training: Working Towards Safer Exercise Options); Andreas Barth, MD, PhD (Gene Therapy in ARVC); Sam Sears, PhD (Fear, Uncertainty and Doubt for ARVC); Hari Tandri, MBBS (Advances in Ablation of Ventricular Arrhythmia in ARVC); Nisha Gilotra, MD (Heart Failure in ARVC: Symptoms, Treatments and Trajectories); Rick Carrick, MD, PhD (The Role of Artificial Intelligence in ARVC); Alessio Gasperetti, MD, PhD (Updates in DSP-Cardiomyopathy); Cindy James, PhD (Research Directions in ARVC-2026 Update). Be sure to check out these amazing presentations using the link below!

The afternoon session offered the opportunity to break out into a variety of patient-led discussion groups, donate blood for our registry, and participate in a Question & Answer Session to close out the seminar. Thank you to everyone who participated in our research studies! We collected 30 blood samples for DNA as part of our ARVC Registry.

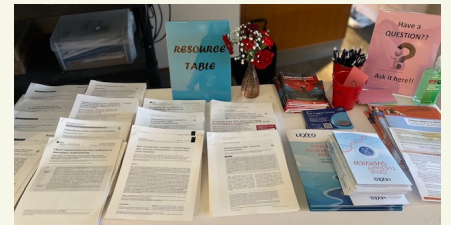
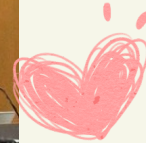
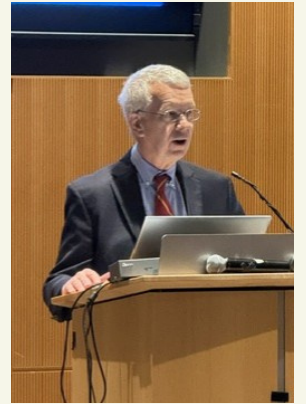
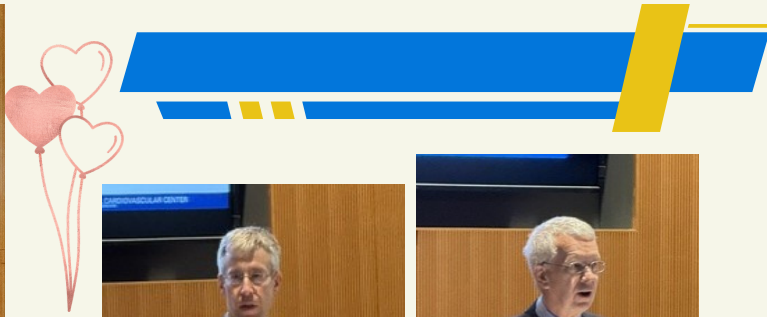
Watch on Demand here: <https://tinyurl.com/ARVCSeminar2026>



*~ The ARVC/C Program
Hugh Calkins, MD
Cindy James, PhD
Brittney Murray, MS
Crystal Tichnell, MGC, RN*



**Family Seminar
SAVE THE DATE!!!
April 24th, 2027**



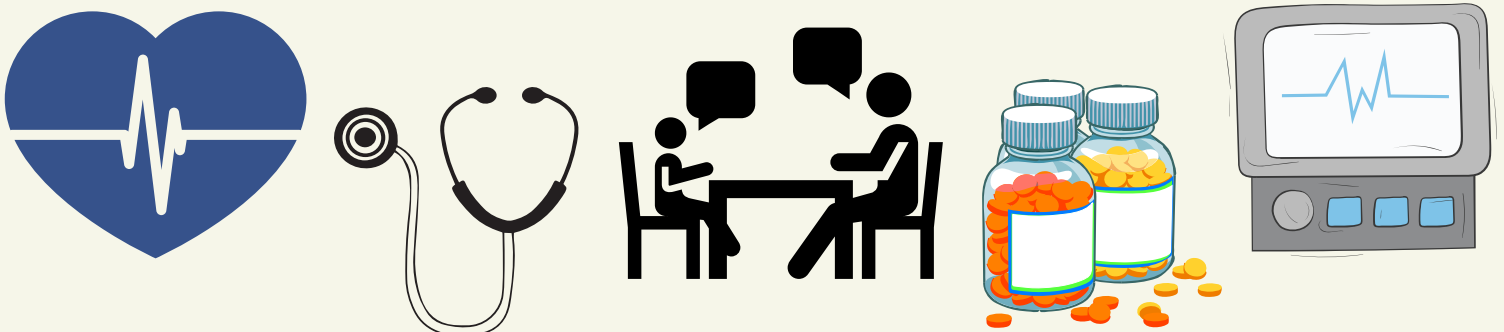
CLINICAL SERVICES AT JOHNS HOPKINS

The Johns Hopkins Arrhythmogenic Cardiomyopathy 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.

New patients are seen in consultation with Dr. Hugh Calkins 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 with Dr. Konstantinos Aronis and Dr. Jonathan Chrispin. Appointments with our heart failure specialist, Dr. Nisha Gilotra, can also be arranged. All appointments are billed to your health insurance.

While all appointments with our physicians are scheduled as in-person visits, there may be some flexibility with our genetic counseling ONLY visits to be scheduled via telemedicine. Please reach out to Crystal to see if you are eligible for a telemedicine appointment based on your appointment needs and physical location. 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 early changes in your health, rather than react to an urgent situation.

To schedule an appointment, contact Crystal at ctichnell@jhmi.edu or 410-955-7292.

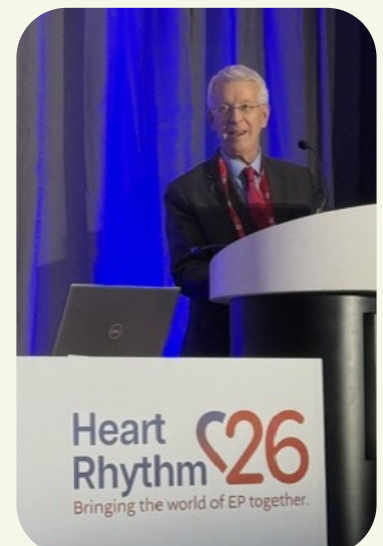


HEART RHYTHM 2026 UPDATE

Chicago, Illinois

The Heart Rhythm (HRS) Society hosted its annual meeting in Chicago this past April (our usual seminar weekend!) and is the largest gathering of heart rhythm professionals from around the world. Several members of our team presented on ARVC and are listed below. It was great to catch up with past fellows and collaborators as well.

- Cynthia James: *New and Evolving Models of Genetics in Cardiology Practice: Results of the RESEQUENCE Trial*
- Brittney Murray: *Accelerating access to guideline-directed genetic test results while maintaining the patient-reported benefits of cardiovascular genetic counseling: Results of RESEQUENCE-GC (NCT05422573)*
- Alessio Gasperetti: *External validation of the DSP-Risk Score for prediction of clinically significant ventricular arrhythmias in patients with desmoplakin cardiomyopathy associated genetic variants*
- Cynthia James: *Arrhythmogenic Cardiomyopathy: A Gene-specific Exercise Prescription for RTP*
- Brittney Murray: *Outcomes Data on Genetic Counseling Independent of Genetic Testing: A MultiCenter Perspective*
- Hugh Calkins: *Debate PRO: Exercise restriction is life saving in pediatric genotype positive, phenotype negative ARVC patients*
- Andreas Barth: *Clinical Outcomes of Gene Therapy in Patients with Inherited Arrhythmias: Insights from Early Cardiac Gene Therapy Trials*





HRS is a great time to meet up with colleagues who have been a part of the Johns Hopkins ARVC journey.



With the "Gene-Based Approach to Arrhythmogenic Cardiomyopathies: An International Expert Consensus Document" now submitted for publication, we turn our focus to the gene elusive patients and had our first brainstorming session at HRS.



2026 Allied Professional and Genetic Counselor Research Award

The SADS Foundation announced the first SADS Allied Professional and Genetic Counselor (AP/GC) Research Award to our very own, Brittney Murray, during the HRS conference! This award recognizes genetic counselors and allied health professionals who conduct outstanding research in cardiac channelopathies and cardiomyopathies. The work should be scientific, innovative, informative, and practical. The topic of the research must be related to inherited channelopathies, inherited cardiomyopathies, or sudden death. It should impact patient care, advocacy, education, research, community awareness, or increase awareness of a new aspect of the disease.

Accelerating access to guideline-directed genetic test results while maintaining the patient-reported benefits of cardiovascular genetic counseling: Results of RESEQUENCE-GC (NCT05422573)

Brittney Murray, MS, CGC, Bryana Rivers, MS, CGC, Katie Nunez, BS, Crystal Tichnell, MGC, RN, Carolyn Applegate, MGC, CGC, Emily Brown, MGC, CGC, Rebecca McClellan MGC, CGC, Andreas Barth, MD, PhD, Casey Overby Taylor, PhD, Jeff Day, MD, MA, Lisa Yanek, MPH, Cynthia A. James, ScM, PhD, CGC

Guidelines emphasize genetic testing (GT) in management of patients with cardiovascular (CV) disease. Supported by data suggesting improved outcomes, genetic counseling (GC) is recommended in conjunction with GT, yet studies suggest providers have difficulty being guideline compliant due to lack of access to CV GC, delaying clinical decision making. Hence, the field is exploring alternatives, such as moving GC to post testing to increase access.

Objective: Test if a post-test model of GC can reduce time from referral to informative GT result while maintaining patient reported psychosocial outcomes.

Conclusion: Prioritizing GC time in a post-test visit while utilizing a genetic education video is an effective way to decrease time to return of medically actionable GT result while maintaining important patient-reported psychosocial outcomes.

Full manuscript has been submitted for review.

Brittney receiving her award from Walker Frahm, CEO of the SADS Foundation, at the 2026 HRS conference in Chicago this past April.





International SADS Foundation Family Conference



November 13th –
November 15th



Texas Children's Hospital
Houston, TX



register at: sads.org/conference2026



FEATURED MANUSCRIPTS



Genome-First Arrhythmogenic Right Ventricular Cardiomyopathy Penetrance Is Not Correlated With Lifetime Exercise History

JACC Adv. 2025 Dec 17;5(1):102464. doi: [10.1016/j.jacadv.2025.102464](https://doi.org/10.1016/j.jacadv.2025.102464)

Eric D Carruth, Weijia Wang, Emily Van Syoc, Catherine Gordon, Christopher D Nevius, Crystal Tichnell, Brittney Murray, Hugh Calkins, Cynthia A James, Christopher M Haggerty

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a heritable cardiomyopathy associated with life-threatening ventricular arrhythmias and is a major cause of sudden cardiac death in the young. Restricting vigorous or endurance exercise is recommended to limit disease progression and sudden cardiac events, and these recommendations extend to genotype-positive, phenotype-negative (G+/P-) individuals.¹ However, the evidence to support these recommendations in the context of genomic screening-based identification (in the absence of personal or family history of disease) remains unresolved.² ARVC penetrance is markedly lower in genomic screening contexts for pathogenic or likely pathogenic (P/LP) desmosome gene variants than disease-based cohorts.³ This presents new management challenges for G+/P- individuals identified through genome sequencing and their providers in establishing physical activity recommendations.

We investigated the role of an individual's lifetime recreational physical activity history in the development of ARVC in the context of a P/LP ARVC-associated desmosome gene variant identified through population genomic screening. To contextualize our genome-screening cohort, we compared our findings with a similar exercise survey of relatives of ARVC-diagnosed probands. We hypothesized that individuals meeting ARVC diagnostic criteria would report higher rates of past participation in recreational exercise, suggesting that physical activity modulates disease expression in those with genetic susceptibility to ARVC.

What is the clinical question being addressed? Does exercise worsen the development of ARVC in genotype-positive individuals from genomic screening?

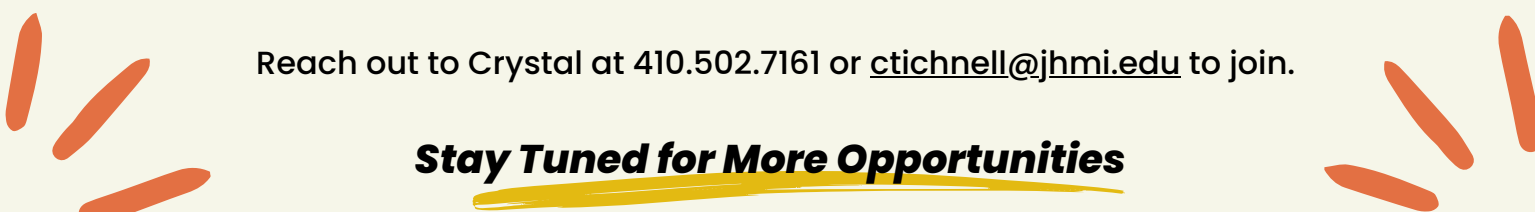
What is the main finding? We find no correlation between exercise history and ARVC development in a genome-screening cohort, contrasting with higher ARVC diagnosis in referrals.



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

If you would like to be a part of studies like the one described above and help to move the field of ARVC forward, consider participating in the Johns Hopkins ARVC 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.



Stay Tuned for More Opportunities

FEATURED MANUSCRIPTS

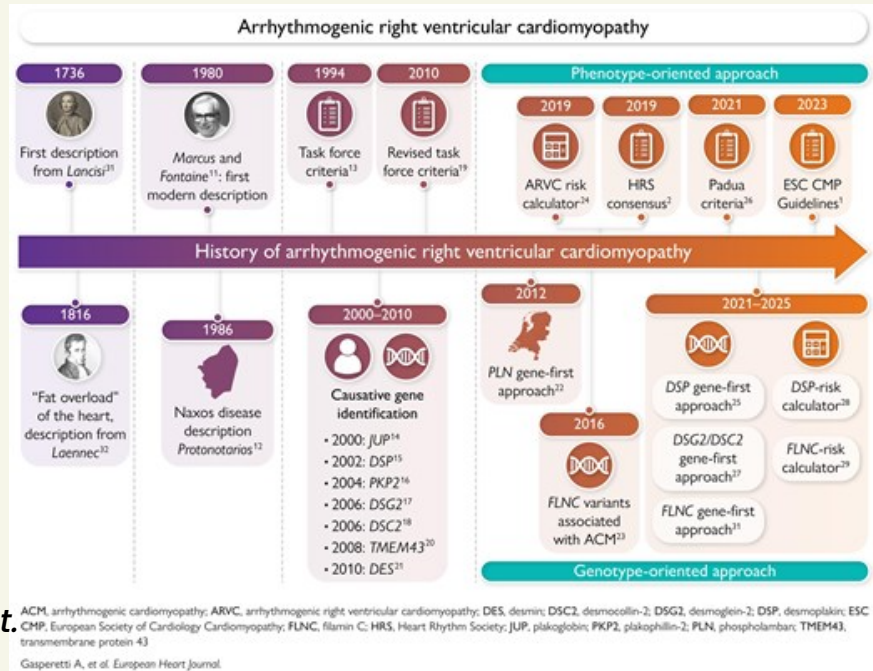
Arrhythmogenic Right Ventricular Cardiomyopathy

Eur Heart J. 2026 Jun 5;ehag297. doi: 10.1093/eurheartj/ehag297. Online ahead of print.

Alessio Gasperetti, Cynthia A James, Firat Duru, Peter van Tintelen, Hugh Calkins

<https://academic.oup.com/eurheartj/advance-article/doi/10.1093/eurheartj/ehag297/8702771>

Arrhythmogenic cardiomyopathy (ACM) is a form of inheritable cardiomyopathy characterized by ventricular scarring and/or fibro-fatty replacement. ACM is associated with a significant risk of sudden cardiac death due to ventricular arrhythmias, especially in young patients and endurance athletes. Aim of this review is to summarize the changing in understanding and framing of this multifaceted disease, in particular highlighting the current transition from a phenotype-first to a genotype-first framework for diagnosis and management.



Alessio Gasperetti



A bittersweet farewell to our research fellow, **Alessio Gasperetti**! We are grateful for his hard work, dedication and friendship. He has made such an impact on DSP research, coordinating the DSPerados network over the past several years. The Johns Hopkins ARVC Program is so thankful to have had Alessio as part of the team for 7 years, publishing over 40 manuscripts on ACM during his time with us. He will be starting his cardiology fellowship at the Cleveland Clinic and we wish him all the best!



WELCOME JILLIAN



Jillian Belluck recently joined the team as our genetic counseling assistant, replacing Anna Nelson. Jillian graduated from Brown University in 2025 with a bachelor's degree in computational biology. While a student at Brown, she was a dedicated teaching assistant for the biology and computer science departments and also participated in research related to malaria and imprinting disorders. After graduating, she spent a year at the Stowers Institute for Medical Research, where she explored complex regions in the genomes of humans, gorillas, and other primates. Jillian plans to pursue a career in genetic counseling and is excited to be supporting the ARVC team!



Moving on

Congratulations to our genetic counseling assistant, Anna Nelson, and our research coordinator, Katie Nunez, as they both move on to graduate school. These two positions are designed as building blocks for young professionals, but saying goodbye is never easy. Anna was often the first point of contact to the program and was responsible for coordinating appointments. Katie worked on the research side and did a lot of data abstraction into our database and making data sheets for our researchers. They both loved to plan the office holiday parties! Anna will be attending the University of Maryland to pursue her master's degree in social work. Katie will be pursuing her master's degree in genetic counseling at Columbia University. We couldn't be more proud of them and are so excited for them to start this next chapter of their careers.



Anna and Katie



Support of the Johns Hopkins ARVC Program Ensures 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 and the team of experts in the ARVC Program are partners in the mission to provide exceptional personalized care, discover better ways to diagnose and treat our patients, and provide educational and training programs for medical professionals, patients, and families. Here are some of the ways that you can help:

Make a Personal Donation

Donations of all sizes, one-time or recurring, make a difference. There are a variety of ways to make a gift to support efforts in the ARVC 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 IRAs, wills and trusts
- Leverage workplace matching gift programs



To make a gift by credit card, visit our online giving form via the QR code

To make a gift by mail, please make a check payable to Johns Hopkins Medicine and indicate the “ARVC Program” on the memo line. Mail to:

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

<https://secure.jhu.edu/form/heart>
Choose “ARVC Program” from the
drop down menu

Launch a Personal Fundraising Campaign

There are many opportunities to become involved in raising awareness and much-needed funds on behalf of the Johns Hopkins ARVC 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 Shannon Brockman,
Associate Director of Development,
at 443-687-2947 or s.brockman@jhumi.edu, for more information.**

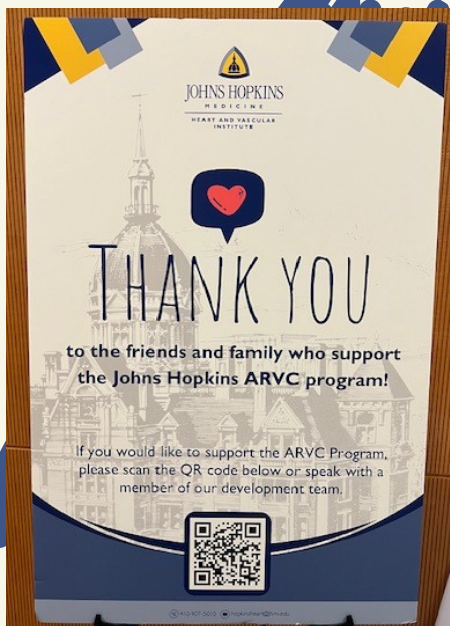
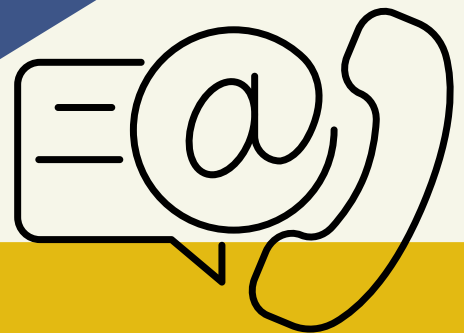
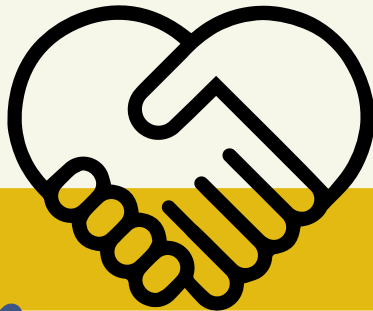
ARVC PROGRAM INFO

ARVC Program Staff

Hugh Calkins, MD—Director
Konstantinos Aronis, MD—Ablation
Jonathan Chrispin, MD—Ablation
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
Anna Nelson—Genetic Counselor Assistant
Katie Nunez—Research Program Coordinator
Leonore Okwara – Research Program Manager
Alessio Gasperetti—Research Fellow

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THANK YOU for your continued generosity and support of the Johns Hopkins ARVC Program. Our program continues to rely heavily on philanthropic funding, now more than ever! Your donations help to support our genetic counselors and research fellows, the core of our program. As the leading ARVC program in the world, we have made great strides in worldwide collaborations to unravel the complexities of Arrhythmogenic Cardiomyopathy, but there is still much work to be done and we can't do it without you!

