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

Winter 2026 *Volume 16, Issue 1*

Happy Holidays from our family to yours!
We wish you good health and new adventures in 2026.

We're excited to host our 27th Annual ARVC/ACM Patient and Family Seminar on Saturday, April 18th, 2026. Unfortunately, due to scheduling limitations, we are unable to offer a hybrid event this year. This year's seminar will be held in-person only.

Additional seminar details can be found inside along with some updates on what we've been working on this past year. Enjoy! Thank you for partnering with us!

2026 ARVC SEMINAR

Presented by
The Johns Hopkins ARVC Program

You and your family members are invited to join us for our annual Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Seminar! This year the seminar will be held in-person only due to scheduling logistics. We apologize for not being able to offer a livestream or recorded option this year. We hope you will consider joining in person to take full advantage of the opportunity to meet other individuals and families, participate in research opportunities, and interact with industry, specifically gene therapy companies.

We are thrilled to have two invited guest speakers, Sam Sears, PhD, cardiac psychologist from East Carolina University, and Adam Helms, MD, cardiologist from University of Michigan with expertise in inherited cardiomyopathies. In addition, we will have presentations from our own Johns Hopkins faculty; Andreas Barth, MD, PhD; Nisha Gilotra, MD; Cindy James, PhD; and Brittney Murray, MS, CGC and special presentations from our research team. You won't want to miss this unique opportunity to meet and network with other families affected by ARVD/C and to learn the latest advances in the field. It will be exciting to share the collaborative efforts all around the world in solving the mysteries of ARVD/C.

Make plans to come into Baltimore early to attend a reception at the Hilton Garden Inn Friday evening (7pm-9pm). Hors d'oeuvres will be served. This event is for patients and family members only. No industry representatives please.

Please register early! There is no registration fee for this seminar, but we continue to be mindful of our gathering capacity. You must register to attend.

WHEN: Saturday, April 18th, 2026 8:00am-5:00pm

COST: Registration is FREE. You just need to get here!

WHO: Patients and Families affected by ARVC, Healthcare Professionals

WHERE: Chevy Chase Conference Center Auditorium
Main level of Sheikh Zayed Tower
1800 Orleans Street
Baltimore, Maryland 21287

REGISTRATION: ALL participants must register! It is also helpful to list names of family members that will be attending with you so we can determine appropriate research opportunities. Register online by 4/1.

https://tinyurl.com/2026ARVCSeminar

Additional Information

HOTEL ACCOMMODATIONS – RESERVE EARLY!!!
Hotel rooms are available (limited) at the Homewood Suites Baltimore Inner Harbor (625 South President Street, Baltimore, Maryland 21202) at a special rate of $179/night plus tax prior to March 18th, 2026 or as long as they are available.

Hotel reservations can be made through the Hilton Garden Inn Central Reservations Line at 888-429-7482. The group name is ARVC Program Seminar Group and the reservation link name is ARVC Program Seminar Group. Reservations can also be made through the online booking link: https://tinyurl.com/arvchotel

The hotel front desk can be reached at 410-234-0999. Check-in 3pm / Check-out 11am. Self-Parking is available for a fee.

TRAVEL TIPS
The Baltimore/Washington International (BWI) Thurgood Marshall Airport is the closet international airport to Johns Hopkins (www.bwiairport.com). It is approximately 30 minutes from the seminar location.

Transportation from Hotel to Seminar – Uber and Lyft are recommended and is at your own expense.

PARKING AT THE SEMINAR
Parking is available at your own expense (max $15) in the Orleans Street Garage. There is a bridge that connects the garage to the main level of Sheikh Zayed Tower (4th floor).

SPECIAL EVENT
Join us for a Meet 'n Greet Reception, 7:00-9:00pm, on Friday, April 17th, 2026 in the Homewood Lodge at the Homewood Suites Baltimore Inner Harbor. Please register for this event when you register for the seminar or contact Crystal. Patients and family members only. No industry representatives please.

CLINIC CONSULTATIONS – REQUEST YOUR APPOINTMENT NOW!!!
Dr. Hugh Calkins and the genetic counselors will be available Friday, April 17th and Monday, April 20th for consultations. Dr. Nisha Gilotra and Dr. Paul Scheel will also have a few clinic slots available. Diagnostic tests can also be arranged if necessary. We ask that if you live locally to please consider arranging your appointment at another time to allow new patients and patients traveling from a distance an opportunity to schedule. These appointments will be billed to your insurance. Please contact Crystal via email at ctichnell@jhmi.edu ASAP to schedule an appointment.

SHARE YOUR STORY
Looking for patient/family stories to share! If you are interested in sharing your story for others to read, please make sure your story and any photos you'd like to include can fit on an 8x10 page. If you've already submitted your story in past years, there is no need to resubmit. We will post your prior submission. Stories need to be submitted to Crystal by April 1st.

QUESTIONS
Contact Crystal Tichnell, MGC, RN at 410-502-7161 or ctichnell@jhmi.edu

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.

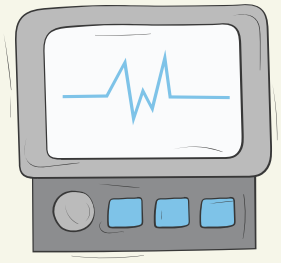
New patients are seen in consultation with Dr. Hugh Calkins and our clinical genetic counselor, Brittney Murray, to discuss test results, family history, and to provide guidance regarding further management. We see all of our patients for genetic counseling to discuss the diagnosis, the psychosocial 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 cardiomyopathy/heart failure specialists, Dr. Nisha Gilotra and Dr. Paul Scheel, 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.







RESEARCH OPPORTUNITIES

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.

Support the Efforts of the Johns Hopkins ARVC Program

Please continue to update your records with us in the registry. Consent to the registry is valid for 5 years and then we require renewal for continued participation. The registry is where all of our research ideas originate and how we achieve long-term understanding of ARVC. Did you know that funding of the registry relies on philanthropy? Your generous donations support our staff and their efforts to get data into the database and out into peer reviewed papers that ultimately change the diagnosis and management of patients with Arrhythmogenic Cardiomyopathies. We need your support and partnership so we can continue making groundbreaking achievements in ARVC.

https://secure.jhu.edu/form/heart?populatedesignation1=ARVC%20Program

Stay Tuned for More Opportunities

FEATURED MANUSCRIPTS

Endurance exercise promotes episodes of myocardial injury in individuals with a pathogenic desmoplakin (DSP) variant

Heart Rhythm. 2025 Nov;22(11):2924-2931. doi: 10.1016/j.hrthm.2024.12.035.

Alan P Jacobsen, Katia Chiampas, Steven A Muller, Alessio Gasperetti, Lisa R Yanek, Richard T Carrick, Catherine Gordon, Crystal Tichnell, Brittney Murray, Hugh Calkins, Lili A Barouch, Cynthia A James.

Background: Desmoplakin (DSP) variants are associated with left predominant or biventricular arrhythmogenic cardiomyopathy. Exercise promotes penetrance and sustained ventricular arrhythmias (VAs) in right-sided arrhythmogenic right ventricular cardiomyopathy, but its effect is unknown in DSP variant carriers.

Objective: The purpose of this study was to assess whether exercise is associated with clinical outcomes in individuals with a pathogenic or likely pathogenic DSP variant.

Methods: Adults with a pathogenic or likely pathogenic DSP variant were interviewed about physical activity from age 10. Endurance athletes were defined on the basis of a mean exercise dose >24 metabolic equivalent hours per week of moderate- to vigorous-intensity exercise. Lifetime survival free of VA (ventricular tachycardia/fibrillation or appropriate implantable cardioverter-defibrillator therapy), clinical heart failure (HF) (presentation to the emergency department or hospitalization with HF), and myocardial injury events characteristic of DSP cardiomyopathy (symptoms, elevated troponin, and imaging with nonobstructive coronaries) were examined using the Kaplan-Meier method and Cox regression models.

Results: Participants (N=100; 66% female; mean age 36 ± 15 years) were active with a median 28.4 (interquartile range 14.8-46) metabolic equivalent hours per week of pre-baseline evaluation exercise, and just 8 individuals continued athlete-level exercise post-baseline evaluation. In multivariable analyses, endurance athletes (60%) had no worse survival free of VA (hazard ratio [HR] 1.00; 95% confidence interval [CI] 0.5-1.98) or clinical HF (HR 0.86; 95% CI 0.36-2.05) but their risk of myocardial injury was elevated (HR 2.37; 95% CI 1.11-5.05). Furthermore, myocardial injury episodes were strongly associated with an elevated risk of both VA (HR 7.86; 95% CI 3.56-17.33) and clinical HF (HR 10.28; 95% CI 2.95-35.83) thereafter.

Conclusion: Endurance exercise may promote progression of DSP cardiomyopathy by increasing the risk of myocardial injury episodes, but the effect on VA and clinical HF is less clear. This study informs shared decision-making exercise and sport participation discussions.

Arrhythmogenic Cardiomyopathy: Towards Genotype Based Diagnoses and Management

J Cardiovasc Electrophysiol.2025 Oct;36(10):2662-2670. doi: 10.1111/jce.16519.

Steven A Muller, Giorgia Bertoli, Jianan Wang, Alessio Gasperetti, Moniek G P J Cox, Hugh Calkins, Anneline S J M Te Riele, Daniel P Judge, Mario Delmar, Richard N W Hauer, Gerard J J Boink, Marina Cerrone, J Peter van Tintelen, Cynthia A James

Arrhythmogenic cardiomyopathy (ACM) is a genetically heterogeneous inherited cardiomyopathy with an estimated prevalence of 1:5000-10 000 that predisposes patients to life-threatening ventricular arrhythmias (VA) and sudden cardiac death (SCD). ACM diagnostic criteria and risk prediction models, particularly for arrhythmogenic right ventricular cardiomyopathy (ARVC), the most common form of ACM, are typically genotype-agnostic, but numerous studies have established clinically meaningful genotype-phenotype associations. Early signs of ACM onset differ by genotype indicating the need for genotype-specific diagnostic criteria and family screening paradigms. Likewise, risk factors for SCD vary by genetic subtype, indicating that genotype-specific guidelines for management are also warranted. Of particular importance, genotype-specific therapeutic approaches are being developed. Results from a randomized controlled trial for flecainide use in ARVC patients are currently pending. Research in a plakophilin-2-deficient mouse model suggests this antiarrhythmic drug may be particularly useful for patients with likely pathogenic or pathogenic (LP/P) PKP2 variants. Additionally, the first gene therapy clinical trials in ARVC patients harboring LP/P PKP2 variants are currently underway. This review aims to provide clinicians caring for ACM patients with an up-to-date overview of the current literature in genotype-specific natural history of disease and management of ACM patients and describe scientific advances that have led to upcoming clinical trials.

Free article can be found here: https://pmc.ncbi.nlm.nih.gov/articles/PMC12530674/


2025 Publications


Muller SA, Asatryan B, Murray B, Tichnell C, Cox MGPJ, Amin AS, Yap SC, Gasperetti A, Carrick RT, Cadrin-Tourigny J, Oerlemans MIFJ, Calkins H, van Tintelen JP, James CA, Te Riele ASJM. **Performance of ARVC Risk Calculators in (Likely) Pathogenic Plakophilin-2 Variant Carriers Without Definite ARVC Diagnosis.** Circ Arrhythm Electrophysiol. 2025 Jan;18(1):e013144. doi: 10.1161/CIRCEP.124.013144. Epub 2024 Dec 13.PMID: 39670315 No abstract available.

Gasperetti A, Carrick RT, Protonotarios A, Murray B, Laredo M, van der Schaaf I, Lekanne RH, Syrris P, Cannie D, Tichnell C, Cappelletto C, Gigli M, Medo K, Saguner AM, Duru F, Gilotra NA, Zimmerman S, Hylind R, Abrams DJ, Lakdawala NK, Cadrin-Tourigny J, Targetti M, Olivotto I, Graziosi M, Cox M, Biagini E, Charron P, Casella M, Tondo C, Yazdani M, Ware JS, Prasad SK, Calò L, Smith ED, Helms AS, Hespe S, Ingles J, Tandri H, Ader F, Peretto G, Peters S, Horton A, Yao J, Dittmann S, Schulze-Bahr E, Qureshi M, Young K, Carruth ED, Haggerty C, Parikh VN, Taylor M, Mestroni L, Wilde A, Sinagra G, Merlo M, Gandjbakhch E, van Tintelen JP, Te Riele ASJM, Elliott PM, Calkins H, James CA. **Clinical features and outcomes in carriers of pathogenic desmoplakin variants**. Eur Heart J. 2025 Jan 21;46(4):362-376. doi: 10.1093/eurheartj/ehae571.PMID: 39288222 Free PMC article.

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Carrick RT, Carruth ED, Gasperetti A, Murray B, Tichnell C, Gaine S, Sampognaro J, Muller SA, Asatryan B, Haggerty C, Thiemann D, Calkins H, James CA, Wu KC. **Improved diagnosis of arrhythmogenic right ventricular cardiomyopathy using electrocardiographic deep learning.** Heart Rhythm. 2025 Apr;22(4):1080-1088. doi: 10.1016/j.hrthm.2024.08.030. Epub 2024 Aug 20.PMID: 39168295

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Muller SA, Bertoli G, Wang J, Gasperetti A, Cox MGPJ, Calkins H, Riele ASJMT, Judge DP, Delmar M, Hauer RNW, Boink GJJ, Cerrone M, Tintelen JPV, James CA. **Arrhythmogenic Cardiomyopathy: Towards Genotype Based Diagnoses and Management**. J Cardiovasc Electrophysiol. 2025 Oct;36(10):2662-2670. doi: 10.1111/jce.16519. Epub 2024 Dec 2.PMID: 39623588 Free PMC article. Review.

Tramèr L, Saguner AM, Beltrami FG, James CA, Calkins H, Duru F. **Gene-elusive arrhythmogenic cardiomyopathy: Roles of sports, inflammation, and beyond**. Heart Rhythm. 2025 Oct 10:S1547-5271(25)02964-9. doi: 10.1016/j.hrthm.2025.09.048. Online ahead of print.PMID: 41077366 Free article. Review.

Falana SL, Kazmouz SG, Iwanski JB, Sarvagalla S, Bas BE, Juneman E, Moukabary T, Ma N, Gundry RL, Rohani L, Hanson P, Laksman Z, James CA, Calkin H, Churko JM. **Modelling arrhythmogenic cardiomyopathy fattyfibro pathology with PKP2-deficient epicardial cells derived from human iPSCs.** Commun Biol. 2025 Oct 27;8(1):1502. doi: 10.1038/s42003-025-08921-z.PMID: 41145823 Free PMC article.

Jacobsen AP, Chiampas K, Muller SA, Gasperetti A, Yanek LR, Carrick RT, Gordon C, Tichnell C, Murray B, Calkins H, Barouch LA, James CA. **Endurance exercise promotes episodes of myocardial injury in individuals with a pathogenic desmoplakin (DSP) variant.** Heart Rhythm. 2025 Nov;22(11):2924-2931. doi: 10.1016/j.hrthm.2024.12.035. Epub 2024 Dec 30.PMID: 39742986

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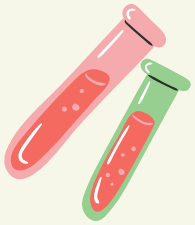
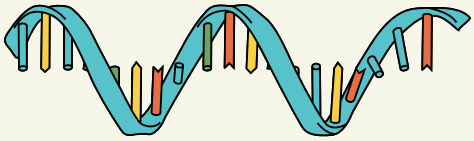
Zhang Y, Prakosa A, Zhang K, Carrick R, Chrispin J, Zimmerman S, Aronis K, Kholmovski E, Tichnell C, Murray B, James C, Calkins H, Trayanova N. Genotype-specific **Digital Twins for Arrhythmia Ablation Targeting in Arrhythmogenic Right Ventricular Cardiomyopathy**. Res Sq [Preprint]. 2025 Nov 5:rs.3.rs-8006874. doi: 10.21203/rs.3.rs-8006874/v1.PMID: 41282123 Free PMC article. Preprint.



Gene Therapy Clinical Trials



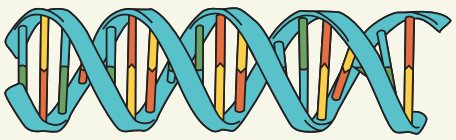
The first in human gene therapy clinical trials have been underway for about a year now. As you consider the question of whether you should participate, it would be prudent to first determine if you meet the strict entry criteria for the study. Current gene therapy trials require patients to be diagnosed with ARVC, have a PKP2 pathogenic variant, and an ICD in place. There are many other inclusion/exclusion criteria as well, which may vary between trials. It is appropriate to weigh options between trials as you decide if participation is right for you.

Assuming you meet enrollment criteria, it is then a difficult personal question whether you should participate. Participation in any study is a personal decision and participation in an early phase “first in man” gene therapy trial is a big decision and a big commitment. Before enrolling in a gene therapy trial, you should have a good understanding of the study goals and take into consideration what the expectations are of you, and what the risks and benefits of the study are before you agree to participate. Gene therapy trials will have strict protocols and it is critical that you adhere to them to ensure the best possible study outcomes. Participation is not for everyone, and that's okay. Each individual has different experiences/circumstances, including varying degrees of symptoms, medications and subsequent side effects, ICD shocks, catheter ablation procedures, etc. that play into how significantly ARVC has impacted their life. Each individual also has different tolerances for risk and different motivations for their decision to participate in an early phase gene therapy trial.

If you are interested and meet the initial eligibility criteria, we are happy to set up a zoom call to discuss gene therapy trials in greater detail. The ARVC Program at Johns Hopkins will be enrolling for both the Tenaya and Lexeo PKP2 Gene Therapy Trials. Email Crystal at ctichnell@jhmi.edu for more information.

Visit: **https://www.clinicaltrials.gov/** for a list of trials currently recruiting.
Consider using search terms such as PKP2, DSP, ACM, ARVC, Arrhythmogenic Cardiomyopathy

We hope this information is helpful as we navigate these new opportunities together.



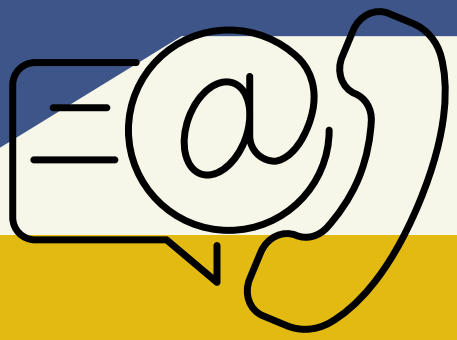
ARVC PROGRAM INFO

ARVC Program Staff

Hugh Calkins, MD—Director
Andreas Barth, MD, PhD - Gene Therapy
Nisha Gilotra, MD—Heart Failure
Paul Scheel, MD—Heart Failure
Konstantinos Aronis, MD—Ablation
Jonathan Chrispin, MD—Ablation
Caridad de la Uz, MD—Pediatrics
Stefan Zimmerman, MD—MR Imaging
Allison Hays, MD—Echo Imaging
Richard Carrick, MD, PhD—Research
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

Contact Us

Johns Hopkins Hospital
600 North Wolfe Street, Blalock 545
Baltimore, Maryland 21287
P: 410-955-7292, F: 443-873-5073
Website: www.ARVD.com
Email: ctichnell@jhmi.edu



Staff Updates

Congratulations to Dr. Alessio Gasperetti as he moves on to the next phase of his cardiology training at the Cleveland Clinic. We will certainly miss him during this time but hope to see him back soon and continue our amazing collaboration on so many important areas of research in ARVC. Dr. Gasperetti has been with the ARVC program for 5 years, earning his PhD, publishing over 50 manuscripts, and leading an amazing international collaboration called the DSPerados Network. We wish him all the best!

JOHNS HOPKINS
MEDICINE

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

Your paragraph text

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.brokcman@jhumi.edu, for more information.