We had a beautiful weekend for this year’s seminar and the most well-attended seminar to date with over 200 in attendance.

Those that arrived into Baltimore early Friday evening attended a Meet ‘n Greet at the Hilton Garden Inn and enjoyed some hors d’oeuvres and company of old and new friends. This was a great opportunity to interact with our staff and share stories with our attendees.

The ABC’s of ARVD/C was standing room only and offered a mini-course on what one needs to know about ARVD/C. This has been a must-attend session for the past 3 years and it will likely be moved to a large meeting space next year! After breakfast, we heard from a number of speakers addressing topics including diagnosis, management, genetics, disease mechanism, drug discovery, catheter ablation, and exercise.

A number of Hopkins faculty members presented including Dr. Hugh Calkins, Dr. Hari Tandri, Dr. Daniel Judge, Dr. Stuart Russell, Dr. Jane Crosson, and Dr. Cynthia James. We were honored to have Professor Andre La Gerche make the 20+ hour flight from Australia to Baltimore to share his work on exercise. We were delighted that Dr. Samuel Sears, PhD from East Carolina University, agreed to skip his year off and return to Baltimore for the 6th time to lead some discussion groups on “Living with Heart” during the afternoon.

In addition, research opportunities were held throughout the afternoon, including blood draws, ICD interrogations, ECGs and Holters, and cheek swab studies. Thank you to everyone who was able to stay and participate in the various research studies. You are a vital part of our research success.
2017 Seminar Date to be Announced Soon!!!

Read what people are saying about the ARVD/C Seminar...

Great Conference! Nice to be able to talk things through with other affected families.

Thank you, thank you, thank you! I am really impressed. I know it was tons of work – gathering info, paperwork, testing, speakers. It touched me that Crystal, Brittney, and Dr. Calkins care about all of us.

The research testing was very well organized and efficient.

The seminar was very informative. It was a pleasure to meet the ARVD staff as well as seeing the attendees again this year.

I can't thank you enough for organizing this seminar. It is serious, comprehensive, and professional. Every field of medicine should do the same thing!

As always, fantastic & educational seminar. Great organization. Thank you for all your years of dedication to ARVD research.

Very informative! I will definitely be coming back yearly for the seminar and recommend for others to come as well.

Amazing seminar!

I had a great time and learned a lot!

It is eye opening how many people do come and that makes it encouraging that we are not alone.

Very useful and informative! A good balance of pragmatic issues and hopefulness!

Thank you for having us. I learned a lot and it was helpful to have the world’s experts on ARVC explain things in terms that were easy to understand.

Did you miss the ARVD/C Seminar this year? Do you still want to learn the latest about ARVD/C? Then click the link below to view the 2016 ARVD/C Seminar Presentations. While definitely not a substitute to the overall Family Seminar experience, you still have the opportunity to view some of the presentations online. Unfortunately, we can’t recreate the special opportunity to interact with the leaders in the field or share personal experiences with other families, a critical aspect to learning how to live with ARVD/C, so start planning now for next year's seminar!

View presentations now at: http://tinyurl.com/2016ARVDSeminar
SAFETY OF AMERICAN HEART ASSOCIATION-RECOMMENDED MINIMUM EXERCISE FOR DESMOSOMAL MUTATION CARRIERS


The goal of this study was to evaluate the role of endurance exercise and exercise intensity with regard to fulfilling Task Force Criteria and development of arrhythmias among family members carrying a PKP2 mutation. In addition, this study assessed whether participation in the American Heart Association (AHA)-recommended minimum for healthy adults (450 to 750 MET-minutes/weekly) was acceptable for at-risk family members. The study population consisted of 28 members of 10 families who had a PKP2 mutation but were not the family proband. They participated in exercise interviews detailing their exercise history from age 10. Results showed that individuals who participated in endurance type and/or higher-intensity exercise were more likely to be diagnosed with ARVD/C. Family members who restricted participation in exercise at or below the upper level of AHA recommended goals were less likely to be diagnosed with ARVD/C. In conclusion, these results suggest that it may be in the best interest of an unaffected desmosomal mutation carrier to refrain from participation in endurance and high-intensity athletics but that participation in the AHA-recommended minimum levels of exercise may be safe. For example, going for a 30-minute brisk walk 5 days per week may be sufficient exercise to not cause additional arrhythmic/progression risks in an ARVD/C patient, but also reduce the risk of developing high blood pressure, high cholesterol, and diabetes.

Clinical Services at Johns Hopkins

The Johns Hopkins ARVD/C Program also provides a variety of clinical services. We see patients for second opinion consultations to discuss diagnosis and management, genetic counseling and testing, as well as 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 one of the genetic counselors 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 specialists, Drs. Stuart Russell, Daniel Judge, and Ryan Tedford can also be arranged. These appointments are billed to your health insurance. To schedule an appointment, contact Crystal.

We are working to expand our clinical services via telemedicine for those who are unable to travel to Baltimore. Our initial launch will consist of genetic counseling services only and eventually expand to second opinion consultations with our electrophysiologists. As soon as details are finalized we will begin to schedule these appointments. If you are interested in telegenetic counseling services only, please email Crystal for additional information and to be added to the waitlist.

We have begun seeing patients in our Pediatric ARVD/C Clinic. This is a twice monthly clinic staffed by a genetic counselor, Brittney Murray, and our specialized pediatric ARVD/C specialist, Dr. Jane Crosson, pediatric electrophysiologist. We offer second opinions/consults for both patients possibly affected and also screening for family history of ARVD/C. This is a coordinated day of completing whatever testing may be necessary along with consults with Brittney and Dr. Crosson. Contact Crystal for information regarding upcoming dates for this special service.

Crystal Tichnell, MGC ctichnell@jhmi.edu 410-502-7161
Heart Rhythm Society Abstract Presentations

The 37th Annual Heart Rhythm's Scientific Sessions were held in Boston in May 2016. The research and collaborative efforts of the Johns Hopkins ARVD/C Program were once again well-represented by several presentations by our team and collaborators.

Starting the meeting off right, ARVD Program genetic counselor Brittnay Murray, MS gave a presentation describing best practices for ordering genetic testing for families with inherited cardiac disease. Hari Tandri, MD discussed how MRI and ablation mapping done during EP studies have clarified the regions of the heart most often involved in ARVC. In her presentation, Cindy James, MS, PhD described how recent research on the role of genotype and exercise help improve management of both patients and families.

On Friday, we had the pleasure of taking part in a session called “Phenotype and the not so obvious genotype” which highlighted new genetic knowledge of ARVD. First, our research collaborator Marina Cerrone, MD from NYU described her work on the overlap of Brugada Syndrome with ARVC. She presented exciting results from a cell system she is using to check whether PKP2 missense variants (single base pair DNA changes) cause problems in heart cells or are just normal variation. We have been selecting variants found in our research participants for her to test. Our previous student, Anneline te Riele, next described our research showing SCN5A mutations and sodium channel dysfunction may plan an important role in ARVD.

On Saturday morning, we joined a dynamic session called “Arrhythmogenic Cardiomyopathy: Where we are and where we’re going” led by our friend, Cristina Basso, MD from Padua Italy. Dr. Calkins first discussed best practices for “Diagnosis and Risk Stratification” stressing the importance of accurate diagnoses, decision-making for defibrillator placement, the importance of limiting data, and presented new data from our team about structural progression in ARVC. Next, Maro Delmar, MD, PhD, from NYU described “Mechanisms: from gene to disease” Dr. Delmar shared that his research team has created a PKP2 mouse model that they are using to better understand the arrhythmias in ARVD as well as what treatment approaches might be most effective in patients.

Recent Publications


Calkins H. *The ventricular ectopic QRS interval for diagnosis and risk stratification in arrhythmogenic right ventricular dysplasia/cardiomyopathy: Is this the answer?* Heart Rhythm. 2016 Apr 5.


What’s New in ARVD/C Research at Johns Hopkins?

GENETICS, MECHANISMS AND CLINICAL PHENOTYPES OF ARRHYTHMOGENIC CARDIOMYOPATHY

The Johns Hopkins ARVD/C Program is participating in a newly funded, multicenter, NIH-sponsored grant as an enrolling center. We are looking for subjects diagnosed with ARVD/C who participated in the previous NIH-sponsored ARVD/C study to re-enroll, as well as their family members. We are also looking for new patients diagnosed with ARVD/C to enroll, along with their family members.

In this study we are trying to find the gene(s) that are responsible for ARVD/C, and to see how the gene(s) affect the onset, the course and the severity of the disease in one individual and/or in a family. Participation will involve sending us your records, yearly follow-up, ECGs, 24 hour Holter monitoring, Signal averaged ECG, 6-minute walk test, and blood donation. In-person visits are required.

If you are interested and want to learn more about your participation and eligibility, please contact Crystal Tichnell, MGC at 410-502-7161 or ctichnell@jhmi.edu.

How Does Family History Influence Psychosocial Adaptation to Inherited Cardiomyopathies?

The goal of this project, sponsored by the National Society of Genetic Counselors 2015 Jane Engelberg Memorial Fellowship, is to determine how family history influences the overall well-being of individuals diagnosed with an inherited cardiomyopathy and their at-risk family members. The first phase of this study has been completed (completing the blue booklet questionnaires) and we are now moving on to the telephone interviews of a subgroup of participants. Thank you for your participation!

The S-ICD – Is it for me?

We are launching a sub-study of our current ICD Registry to assess the efficacy of the new sub-cutaneous device or S-ICD in patients with a diagnosis of ARVD/C. If you have been diagnosed with ARVD/C and had your ICD (transvenous or subcutaneous) implanted after January 2013, you are eligible to participate. Participation involves:

1) Enrollment in our ARVD/C Registry
2) Sending us medical records
3) Sending us interrogations from your device
4) Contact us if you have an ICD therapy, ICD shock, ICD replacement, or other procedure related to your device.
5) Completion of questionnaires.

Email Crystal at ctichnell@jhmi.edu to discuss your eligibility and enrollment.
Clinical and Genetic Investigations of Right Ventricular Dysplasia (ARVD/C Registry)

Who: Children and adults with ARVD/C

What: Collection of pertinent past medical records and continued collection for 5 years. A blood sample for DNA for genetic mapping of ARVD/C genes

How to Join: Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu. She will need to send you a consent form, then review the submitted records and make arrangements for obtaining and shipping the blood sample.

* * * * *

Have you had an epicardial ablation?

We are looking for people with ARVD who have had an epicardial ablation to join our Registry. Help us discover how this new technique affects the course of ARVD/C! Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu.

* * * * *

THANK YOU FOR YOUR PARTICIPATION IN ALL OF THESE IMPORTANT STUDIES!!!

Clinical Updates - Genetic testing and Exercise

Genetic Testing Updates

Have you had Genetic Testing that was Negative?

Genetic testing has advanced tremendously over the past decade. At one point, there was only screening for a single gene, PKP2 or plakophilin-2. Now, there are panels consisting of 100 various genes that have been linked to arrhythmias and cardiomyopathy. If you have had prior genetic testing that only looked at a handful of genes and was negative, talk to your genetic counselor and/or physician about the option of additional genetic testing looking at a broader range of genes. We are finding that not all cases of ARVD/C are caused by desmosomal genes and have branched out looking at other possible causative genes.

Exercise Recommendations

Over the past several years, our research has been focused on understanding the impact of exercise on those diagnosed with ARVD/C, as well as with those who are at increased risk of developing ARVD/C (ie. carry a genetic mutation, but don’t have evidence of disease). There is increasing evidence that participation in competitive endurance types of intense exercise acts as a trigger for progression and risk of sudden cardiac death in ARVD/C. We have also shown that those who do not yet have an identifiable genetic cause for their ARVD/C tend to be even more athletic in nature and require more exercise than those who do have an identifiable genetic cause. We were honored to have Dr. Andre La Gerche present at this year’s ARVD/C seminar and share his understanding of the athlete's heart and the impact of intense extreme exercise. It was mentioned at the seminar that weight lifting may not have as big of an impact on the RV as once previously thought. However, this has not been formally studied and our recommendation remains that only light to moderate weight lifting may be appropriate and requires additional research to confirm and clarify. It is extremely important to discuss with your physician about what types of exercises are most appropriate for your specific case.
Diagnosed with ARVD/C in April of 2015 after a v-tach event during swim practice, Blanchard Seniff has been eager to raise funds to support ARVD research. Teenage friends from the Coronado, California community rallied around Blanchard and created this video and coordinated several fund raising events. Students organized into “Team Blanchard” and set up a Team Blanchard for ARVD FaceBook page to raise awareness, and a GoFundMe page: https://www.gofundme.com/teamblanchard to see their progress online along with a Team Blanchard tracking account with the Johns Hopkins ARVD program. Together so far, Team Blanchard has raised over $30,000 for ARVD research.

Team Blanchard launched its fund raising efforts first through a social media blitz which generated several grass roots efforts to raise additional funds. Friends and community members held a “Yoga on the Beach” event, several restaurants sponsored a “Restaurant week for ARVD”, a local surf wear t-shirt company allowed Blanchard to adapt their logo and design a T-shirt with all proceeds going to Johns Hopkins University ARVD Program. The mayor of Coronado went on to issue a City Proclamation naming February 2016 Coronado Heart Month and the local Fire Department and Paramedic teams supported Team Blanchard events educating the public on AED use and cardiac event response. The Coronado Rotary Club, Coronado High School and the Coronado Middle School all sponsored Team Blanchard, raising funds and creating additional video news broadcasts which showcased how to use an AED, and respond to a cardiac event. Team Blanchard is well on its way to meet its goal of $100,000 for Johns Hopkins ARVD Research. Blanchard is grateful for everyone's support and the opportunity to change the future for people affected by ARVD. Go find that cure! ~ The Seniff Family

Listen to Blanchard’s story here: https://vimeo.com/153565786

THANK YOU TO THE SENIFF FAMILY FOR ALL OF YOUR EFFORTS IN RAISING FUNDS AND INCREASING AWARENESS FOR ARVD/C.
How to Support our ARVD/C Program Efforts

Thank you for your interest in making a charitable gift, fundraising philanthropic support and/or building awareness for the Johns Hopkins ARVD/C Program! There are many ways in which you can support our ARVD/C Program efforts, which we have listed below for your convenience:

- **Make a charitable contribution** to support ARVD/C research by phone at 443-287-7382 or online at [http://www.arvd.com/donations.html](http://www.arvd.com/donations.html) or by mail to our Development Office listed below.
- **Consider making bigger impact** by making a monthly gift or multi-year pledge that is easy, efficient, and helpful to the ARVD/C faculty and staff for future planning.
- **Honor or memorialize someone special** by making a gift to Johns Hopkins ARVD/C Program in his/her name.
- **Fuel future ARVD/C efforts** by including the Johns Hopkins ARVD/C Program in your will or trust, or designating it as a beneficiary of a retirement account or life insurance policy.
- **Ask your friends and family members** to contribute to the ARVD/C Program.
- **Ask your company** to match your contributions.
- **Contribute a portion of your company’s sales** to the ARVD/C Program and/or invite customers to directly contribute by posting a link to [www.arvd.com](http://www.arvd.com) on your website.
- **Raise funds and awareness** for the ARVD/C Program via social media.
- **Create pillows, tee-shirts, etc.** and use the proceeds to support the ARVD/C Program.
- **Hold a personal fundraising event or auction** at your school, a local health fair, your yoga studio, your home, your favorite restaurant, etc. and donate the proceeds to the ARVD/C Program. (We would like to learn about your fundraising event! Please contact our Development Office below to get started.)
- **Share your unique story** and provide referrals to Johns Hopkins ARVD/C Program.

We welcome your questions, concerns, ideas and feedback! Please contact **Molly C. Dolan** at the Heart and Vascular Institute Development Office for more information:

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**ARVD/C Program Info**

**ARVD Program Staff**

Hugh Calkins, MD—Director  
Harikrishna Tandri, MD—Faculty  
Daniel Judge, MD—Faculty  
Stuart Russell, MD—Faculty  
Theodore Abraham, MD—Faculty  
Gabriela Orgeron, MD—Post Doctoral Research Fellow  
Aneline te Riele, MD—Post Doctoral Research Fellow  
Cynthia James, ScM, PhD—Genetic Counselor  
Brittney Murray, MS—Genetic Counselor  
Crystal Tichnell, MGC—Genetic Counselor  
Bryana Rivers—Genetic Counselor Assistant

**Support Group Info**

Looking for a support group?

**ARVD support group on Google:**  
Search for "ARVD ARVC Support Group" on google groups. Any issues joining this group, email Bob at ralla52@yahoo.com

**FACEBOOK Groups:**  
- **ARVD/C Youth Society** – private group on Facebook (request invite from group admin)

- **Hope for ARVD** – private – request access

- **The Broken Heart Club – ARVD Edition** – private – request access

**ARVD/C Mentor Program:**  
Get matched with an ARVD/C mentor! Connect with a mentor who has navigated the challenges of life with ARVD/C and receive: Support, Connection, Understanding, and Strategies for Thriving with ARVD/C. Contact Nancy Bogle at nbstjohn@gmail.com for more information and visit ARVDHEARTANDSOUL.ORG

Don’t forget to keep us informed of your most up-to-date contact info!  
Please send any changes and updated medical records to Crystal at ctichnell@jhmi.edu  
Thank you!

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www.ARVD.com