WELCOME 2016!! From all of us here at the ARVD/C Program, we hope you and your families have had a wonderful Holiday Season. We wish you good health, happiness, and success in the coming year and always! This past year the ARVD/C Program has continued working with collaborators from around the world. We’ve wrapped up some projects and launched some new ones so once again we’ll be looking for your help.

Please know that your continued support both financially as well as through your research participation does not go unnoticed. Thank you for volunteering your records, your time, and your ideas. Advances that have been made in the field of ARVD/C are all because of your dedication. We cannot thank you enough! We are excited to announce our Annual Patient and Family Seminar this Spring and hope to see many of you there. Details are enclosed. Please do not hesitate to contact our program with any questions or concerns.

~ The ARVD/C Program

Clinical Services at the ARVD/C Program

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 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 at 410-502-7161 or ctichnell@jhmi.edu.

The world’s only specialized Pediatric ARVD/C Clinic began this past Fall. This will be a twice monthly clinic staffed by a genetic counselor, Brittney Murray, and our specialized pediatric ARVD/C specialist, Dr. Jane Crosson, pediatric electrophysiologist. We will offer second opinions/consults for both patients possibly affected and also cardiac testing associated with screening for family history of ARVD/C. Contact Crystal for information regarding upcoming dates for this new special service.

Planning to come to the seminar in April??

Would you like to schedule a consultation with the ARVD/C Team to discuss your diagnosis, management, and/or genetic testing??

→ Then request your appointment NOW! ←

We will be available for consultations on Friday, April 29th AND Monday, May 2nd. These appointments go quickly. Your insurance company will be billed for these services. Contact Crystal @ 410-502-7161 or ctichnell@jhmi.edu to request an appointment.
You and your family members are invited to join us for our annual ARVD/C Seminar! You won’t want to miss this unique opportunity to meet other families affected by ARVD/C and to learn the latest advances in the field. We are thrilled to have a number of Johns Hopkins faculty members present this year, in addition to several guest speakers. Professor André La Gerche, MBBS, PhD, from Australia, will share his work on the effect of endurance exercise on the heart and Dr. Samuel Sears, cardiac psychologist from East Carolina University in Greenville, North Carolina will make a return appearance. After lunch, there will be an opportunity to ask questions in a “Question and Answer Session with the Experts”, as well as an “Under 30” Discussion Group and opportunities to participate in research. Make plans to come into Baltimore early to attend a reception at the Hilton Garden Inn Friday evening. Once again, we have an exciting and packed agenda so we look forward to seeing many of you in April! Please register early! There is no registration fee for the seminar. All are welcome!

WHEN:   Saturday, April 30th, 2016 8:00am-5:00pm

WHO:     Patients and Families affected by ARVD/C, Healthcare Professionals

WHERE:   Chevy Chase Conference Center Auditorium, main level of Sheikh Zayed Tower at 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 April 15th.


HOTEL ACCOMODATIONS – RESERVE EARLY!!!
Hotel rooms are available (limited) at the Hilton Garden Inn, Baltimore Inner Harbor (625 South President Street, Baltimore, Maryland 21202) at a special rate of $149/night plus tax (single/double) until March 29th, 2016 or as long as they are available. Call 1-877-STAY-HGI or 1-877-782-9444 and mention the “ARVD Group room block rate” to receive the special rate. Check-in 3pm / Check-out 12pm. Hotel front desk phone is 410-234-0065. Self-Parking is available at a rate of $23 and valet $32 per day. www.baltimoreinnerharbor.hgi.com
Reservations can also be made online using this link: http://hiltongardeninn.hilton.com/en/gi/groups/personalized/B/BWIIHGI-RVD-20150429/index.jhtml?WT.mc_id=POG

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

A bus will be available to transport seminar participants to the seminar on Saturday, departing only from the Hilton Garden Inn at 7:30am with a 2nd bus leaving at 8am. The bus will leave the seminar at 5:00pm to return to the Hilton Garden Inn.

Taxi Services – For Taxi services call Sun Cab at 410-235-0300 or Yellow Cab at 410-752-1096.

PARKING AT THE SEMINAR
If you are not taking the bus from the hotel, parking is available at your own expense (max $12) in the Orleans Street Garage. There is a bridge that connects the garage to the main level of Sheikh Zayed Tower.

SPECIAL EVENT
Join us for a Meet ‘n Greet Reception, 7:00-9:00pm, on Friday, April 20th, 2016 in the Great American Grill at the Hilton Garden Inn. H’ors deurves will be served. Please register for this event when you register for the seminar or contact Crystal.

CLINIC CONSULTATIONS
Dr. Hugh Calkins and the genetic counselors will be available both Friday, April 29th and Monday, May 2nd for consultations. Diagnostic tests can also be arranged if necessary. These appointments will be billed to your insurance. Please contact Crystal ASAP to schedule an appointment.

RESEARCH
Research opportunities will be available on Saturday afternoon and will include blood drawing, ICD interrogations, etc. We must have your medical records ahead of time to determine if you are eligible for research.

QUESTIONS
Contact Crystal Tichnell, MGC at 410-502-7161 or ctichnell@jhmi.edu
The American Heart Association’s Scientific Sessions is the leading annual convention for scientists and healthcare professionals devoted to the science of cardiovascular diseases and stroke, and the caring of patients suffering from these diseases. Its mission is to build healthier lives, free of cardiovascular diseases and stroke. The American Heart Association Scientific Sessions were held in Orlando, Florida in Nov 2015. Four abstracts were presented at this year’s conference and are summarized below.

**CHARACTERIZING THE MOLECULAR PATHOLOGY OF ARRHYTHMOGENIC CARDIOMYOPATHY IN BUCCAL MUCOSA CELLS**
Angeliki Asimaki, Cynthia A James, Stephen P Chelko, Crystal Tichnell, Brittney Murray, Adalena Tsatsopoulou, Alexandros Protonotarios, Anneline te Riele, Andre G Kleber, Daniel P Judge, Hugh Calkins, Jeffrey E Saffitz

The purpose of this study was to evaluate the use of buccal mucosa (cheek) cells (as opposed to actual heart cells) in the evaluation of protein expression. Buccal mucosa smears from 57 subjects (40 clinically diagnosed ARVC patients, 5 mutation carriers without disease, and 12 controls) were immunostained in a blinded fashion. Junctional signal for various proteins which are decreased in heart muscle of ARVC patients, were also reduced in buccal cells of most ARVC patients. Due to the readily accessible nature of cheek cells and the fact that they can be cultured, disease mechanisms and disease modifiers can be more easily studied. Thank you to all who donated cheek cells for this research!

**CLINICAL PRESENTATION, CARDIAC PHENOTYPE AND LONG TERM PROGNOSIS OF PATIENTS WITH LATE ONSET ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY**
Aditya Bhonsale, Anneline te Riele, Abhishek Sawant, Judith Greneweg, Cynthia A James, Brittney Murray, Crystal Tichnell, Dennis Dooijes, Jeroen vd Heijden, Harikrishna Tandri, Peter van Tintelen, Daniel P Judge, Richard Hauer and Hugh Calkins

This study describes the phenotypic features and clinical course of those ARVC patients presenting after the age of 50. This late presentation was observed in 21% of patients participating in the Johns Hopkins and Utrecht ARVC registries. The most common presenting symptom was sustained ventricular tachycardia (VT). They were less likely to experience syncope or have a pathogenic mutation. They also had few numbers of PVCs on their Holter monitors compared to those diagnosed before the age of 50 years. Late presentation does not necessarily result in a benign course.

**HEART FAILURE IS COMMON AND UNDER RECOGNIZED IN PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY**

As improvements have been made in the diagnosis of ARVC and prevention of sudden cardiac death with ICDs, heart failure (HF) is becoming more prevalent amongst ARVD/C patients. 288 ARVC patients were assessed for HF signs and symptoms. 236 had evidence of structural heart disease defined as right ventricular (n=199) or biventricular dilatation/dysfunction (n=37). Of those with identified structural disease, 52% of patients reported at least one clinical sign or symptom of HF with dyspnea on exertion and fatigue being the most common. Edema was found in 20% of patients. Sixteen patient underwent heart transplant. HF symptoms appear to be more common in the ARVC population than originally thought and may be under recognized.

**PREDICTIVE VALUE OF LOCAL PROLONGED ELECTRO-MECHANICAL INTERVAL IN THE CONCEALED STAGE OF ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY**
TP Mast, AJ Teske, AS te Riele, JA Groeneweg, C Tichnell, B Murray, H Tandri, JF van der Heijden, BK Velthuis, P Lah, PA Doevendans, D Dooijes, TA van Veen, JM de Bakker, TP Abraham, CA James, H Calkins, RN Hauer, MJ Cramer

This study sought to utilize electro-mechanical interval (EMI) as a potential new parameter in unmasking activation delay in the early stages of ARVD/C. Mutation positive definite ARVD/C patients (n=108), asymptomatic mutation carriers not fulfilling task force criteria (n=35), and healthy controls (n=50) underwent echocardiographic deformation imaging and an electrocardiogram. EMI was prolonged in all right ventricular segments measured in ARVD/C patients. EMI was prolonged in the subtricuspid region of 17/35 asymptomatic mutation carriers. In follow-up, 11/35 mutation carriers experienced an increase in ventricular arrhythmias. Prolonged EMI may be helpful in the risk stratification of asymptomatic mutation carriers.
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 asking people 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.

Relative Safety, Efficacy, and Patient Satisfaction of Standard ICDs versus the Sub-Cutaneous ICD

Who can participate? Patients diagnosed with ARVD/C and have an S-ICD

What do I have to do? Contact Crystal (ctichnell@jhmi.edu) to enroll in the study. You will need to sign a consent form, send us your medical records, and complete online questionnaires.

Do I have to travel to Johns Hopkins? No

Sponsored by Boston Scientific
PI: Hugh Calkins, MD
Johns Hopkins IRB NA_00042471 (Predictors of Implantable Cardioverter Defibrillator (ICD) Firing in Right Ventricular Dysplasia)
Ongoing Research Opportunities at Johns Hopkins

Clinical and Genetic Investigations of Right Ventricular Dysplasia (Registry)

Who: Children and Adults with ARVD

What: Collection of pertinent past medical records and continued collection for 5 years. A blood sample for DNA for genetic mapping of ARVD genes will also be collected.

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! Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu

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

UPDATE: 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. Thank you to everyone who participated by either completing an online questionnaire or the blue book questionnaire. Some of you may be contacted to complete a telephone interview shortly. We are busy analyzing all of the data and will report back soon!

How You Can Help

None of the research by the ARVD/C Program would be possible without the active participation of families affected by ARVD/C. To join our research, the first step is to enroll in the ARVD/C Registry and send us copies of your cardiac tests. We will then invite you to be a part of other research efforts that are appropriate for you.

While we continue to apply for funding to support our various research projects, we still rely heavily on the generosity of families to financially support this program.

If you are interested in making a financial contribution to the ARVD/C Program, please contact Molly Dolan or Shannon Wolman with our Development Office.

Molly Dolan, Senior Associate Director of Development
Phone: 443-287-7382
Email: mdolan6@jhmi.edu

Shannon Wollman, Director of Development
Phone: 443-287-7383
Email: swollma3@jhmi.edu

You can also make an Online Gift through our encrypted, secure server at: http://www.hopkinsmedicine.org/heart_vascular_institute/clinical_services/centers_excellence/arvd/about/donate.html

If you are hosting an ARVD/C Fundraiser and would like to include information regarding your event in our newsletter, please email Crystal at ctichnell@jhmi.edu.
Pregnancy course and outcomes in women with arrhythmogenic right ventricular cardiomyopathy


We had the pleasure of hosting Anke Hodes, a senior medical student from the University Medical Centre Groningen in the Netherlands. Anke joined us from September 2014-January 2015 to do an honors research clerkship – the last step of her medical school training. Anke worked diligently on a project the Hopkins Genetic Counselors have been longing to put together. We are thrilled to present another finished product of the Dutch/North American Collaboration that will hopefully provide physicians with important information they need to care for their ARVC patients considering pregnancy.

The full article is available via the link provided below.

The purpose of this project was to characterize the pregnancy course and outcomes in women diagnosed with ARVD/C. Cardiac symptoms, treatment, episodes of sustained ventricular arrhythmias and heart failure were assessed, as well as obstetric outcomes. We identified 26 women diagnosed with ARVD/C during 39 pregnancies (1-4 per woman). The comparison group consisted of 117 childbearing-aged female patients diagnosed with ARVD/C who had not experienced any pregnancies.

Treatment during the 39 pregnancies consisted of beta blockers (n=16), antiarrhythmics (n=6), diuretics (n=3), and ICDs (n=28). Among 5 pregnancies, the mothers experienced a single sustained ventricular arrhythmia, 2 of which were terminated by the ICD. Heart failure developed in two pregnancies in women who had already been previously diagnosed with biventricular or right ventricular disease. All infants were born without any major obstetric complications. Delivery by caesarean section occurred in 11 pregnancies and were done for obstetric indications with the exception of one (HF). Neither sustained ventricular arrhythmias nor heart failure incidence were significantly increased during pregnancy. The course of ARVD/C did not differ between women who underwent pregnancies and those who did not. In summary, most pregnancies in women with ARVD/C are tolerated well.

Read the full article now!

http://heart.bmj.com/cgi/rapidpdf/heartjnl-2015-308624?ijkey=qon5vA3zf8vGqLT&keytype=ref
**ARVD Program Info**

**ARVD Program Staff**
- Hugh Calkins, MD—Director
- Harikrishna Tandri, MD—Faculty
- Daniel Judge, MD—Faculty
- Stuart Russell, MD—Faculty
- Ryan Tedford, MD—Faculty
- Theodore Abraham, MD—Faculty
- Gabriela Orgeron, MD—Post Doctoral Research Fellow
- Anneline 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.

**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 via Fax: 410-502-9148 or email ctichnell@jhmi.edu** You can also mail records as well. Thank you!

**THANK YOU!!!**
A special thank you to everyone who has supported the Johns Hopkins ARVD/C Program over the past decade. We truly appreciate the generosity of patients and families who have been able to support our program financially. Your donations support our genetic counselors and allow our program to be the forefront of ARVD/C research. We cannot thank you enough for your continued participation in responding to surveys, sending medical records, providing blood/skin samples, etc. Together, we will continue to solve the mysteries of ARVD/C.

**Contact Us**
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