



THE JOHNS HOPKINS HOSPITAL

CENTER FOR INHERITED HEART DISEASE

NEWSLETTER

The Johns Hopkins Center for Inherited Heart Disease is dedicated to providing patients and families with valuable insights into cardiac genetics, promoting awareness, and providing patient education. We hope this newsletter can offer you timely and relevant information to empower you and your loved ones on the path to navigating your heart condition. In each edition, we will delve into various aspects of inherited heart diseases, including:

- **Education and Awareness**: Our goal is to empower you with the knowledge needed to make informed decisions about your health and the health of your loved ones.
- **Patient Stories**: We will feature inspiring stories of individuals and families who have faced the challenges of inherited heart diseases with resilience and determination.
- **Research and Innovation**: We will provide updates on the latest advancements in the field of inherited heart diseases, including breakthroughs in genetic testing, emerging treatment modalities, and ongoing research efforts.
- **Philanthropy and Giving Back**: We will share opportunities for you to get involved and make a difference. Your philanthropic support plays a vital role in furthering our mission, and we deeply appreciate your consideration.

Thank you for reading the Johns Hopkins Center for Inherited Heart Disease Newsletter! As we approach the holiday season, we want to use this opportunity to provide a brief update on what we've been up to over the past couple of years. We continue to strive to learn as much as we can to improve patient care and build community with the patients we serve.

Recent Publications

Multidisciplinary Teams in Clinical Lipidology and Cardiometabolic Care: A National Lipid Association Expert Clinical Review

Mary Katherine Cheeley, PharmD, CLS, FNLA · Carol F. Kirkpatrick, PhD, MPH, RDN, CLS, FNLA · Emily E. Brown, MGC, CGC · Dave L. Dixon, PharmD, CLS, FNLA · Heather Gunter, MSN, MBA, RN, NE-BC · Diane S. Osborn, ANP, CLS, FNLA · Michael J. Wilkinson, MD, FNLA

This National Lipid Association Expert Clinical Review (ECR) describes the roles of multidisciplinary care team members in optimizing the management of lipid disorders and promoting cardiometabolic health, including the contributions of physicians, advanced practice providers, pharmacists, registered dietitian nutritionists, nurses, and genetic counselors. Patients benefit from a collaborative approach to the management of dyslipidemias and cardiometabolic risk through improved diagnosis and treatment, access to comprehensive and evidence-based lifestyle interventions, optimization of pharmacotherapy, and patient education and empowerment. Certain multidisciplinary team members may not be readily accessible in all practice settings. Therefore, this ECR provides suggestions for accessing community resources to expand the reach of multidisciplinary care to a greater number of patients.



External validation of atrial fibrillation risk prediction tools in a racially diverse cohort of patients with hypertrophic cardiomyopathy

Henry Pronovost MD, Ali Asghar Kassamali XX, Jeff Ehmsen MD, PhD, Bryana Rivers MS, CGC, Alessio Gasperetti MD, PhD, Katherine C. Wu MD, Ethan Rowin MD, Jose A. Madrazo MD, Richard T. Carrick MD, PhD



Background

Atrial fibrillation (AF) is the most common sustained arrhythmia in patients with hypertrophic cardiomyopathy (HCM). The 2024 American Heart Association/American College of Cardiology guidelines recommend validated clinical tools such as the HCM-AF score for individualized assessment of AF risk. To date, these tools have been validated only in predominantly white HCM patient populations.

Objective

This study aimed to compare the performances of published AF risk prediction tools in a racially diverse cohort of patients with HCM.

Conclusion

The HCM-AF score demonstrated good AF risk discrimination in a diverse cohort of patients with HCM, outperforming alternative non-HCM-specific AF risk scores and validating its use in nonwhite HCM populations.

Is it time for a paradigm shift? Inclusion of APOE on genetic dyslipidemia panels

Hannah E Ison, Mohammad Mowaswes, Ronen Durst, Thorsten Leucker, Joshua W Knowles, Emily E Brown

APOE codes for apolipoprotein E (ApoE), which plays an important role in lipid and lipoprotein metabolism and homeostasis of tissue lipid content. Several variants in APOE have been associated with inherited dyslipidemias, and a subsequent increased risk of developing premature coronary artery disease (CAD). However, these variants and their impact on risk can be thought of on a spectrum, with some being more monogenic in nature, and others contributing in a polygenic/multifactorial manner. Despite these known associations, there is often

hesitancy around ordering APOE genetic testing due to the association with Alzheimer's disease. This paper aims to catalyze discussion around APOE testing and counseling strategies, highlight the nuances around this topic, and advocate for inclusion of APOE testing on dyslipidemia panels when an inherited dyslipidemia is suspected.



Find out more on
the next page
about the podcast
episode where
Emily discusses
this paper!

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Tune into the DNA Dialogues Podcast to hear our very own, Emily Brown share her expertise. The episode is titled **Shifting Paradigms: APOE in Dyslipidemia & Patient Voices in Neurogenetics**. In the first segment of the episode, Emily and her Stanford colleague, Hannah Ison, discuss their recent research publication regarding the inclusion of the *APOE* gene on dyslipidemia panels. Several variants/mutations in the *APOE* gene have risk associations for dyslipidemia (abnormal lipid levels) and coronary artery disease. However, the *APOE* gene also provides risk information for Alzheimer's disease. For this reason, many ordering providers have been hesitant to include the *APOE* gene when ordering genetic testing for inherited dyslipidemias. Tune in to hear why it may be a good idea to change our approach!



NEW
EPISODE

Optimizing Care for Hypertrophic Cardiomyopathy

A White Paper from the HCM Initiative of
AfPA's Cardiovascular Disease Working Group

Last March, one of our genetic counselors, Bryana Rivers, was invited by the Alliance for Patient Access (AfPA) to join the newly established Hypertrophic Cardiomyopathy (HCM) Initiative within their Cardiovascular Working Group. The meeting was a two day conference in Washington D.C. where several HCM-specialized clinicians discussed the challenges that many of our patients face with access to care. The policy team at AfPA took detailed notes and published an informative white paper called, "Optimizing Care for Hypertrophic Cardiomyopathy" which can be found on their website.

Conclusion

To optimize care for hypertrophic cardiomyopathy, patients need access to the full range of treatment options. Implementing best practices, improving patient education and enhancing collaboration can all enable patients to access the right treatment and pursue better health outcomes.

MEMBERS OF AFPA'S CARDIOVASCULAR DISEASE WORKING GROUP HCM INITIATIVE

Richard Bach, MD	Dharmesh Patel, MD
Matthew Martinez, MD	Bryana Rivers, MS, CGC
Srihari Naidu, MD	Jeffrey Towbin, MD

JHM HCM PATIENT SYMPOSIUM

September 13th 2025



The Johns Hopkins Hypertrophic Cardiomyopathy Center of Excellence hosted a free patient symposium in September of 2025. This event was an opportunity for patients and their families to learn about the latest in HCM treatment and research, as well as to connect with other patients with HCM.

Save the Date for next year's HCM Symposium!



In September 2024 a couple of our genetic counselors attended the National Society of Genetic Counselors Annual Conference, which was in New Orleans, Louisiana this year. The conference is a great opportunity to learn about updates in clinical genetics across multiple specialties and to present research to colleagues across the U.S., Canada, and more!

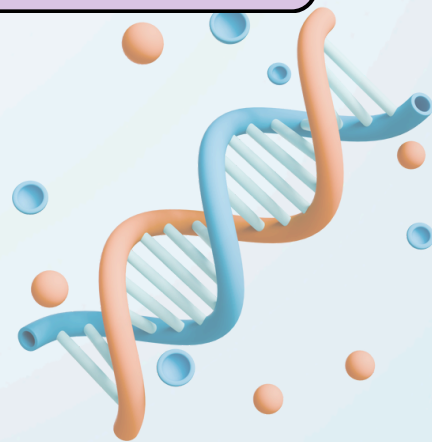
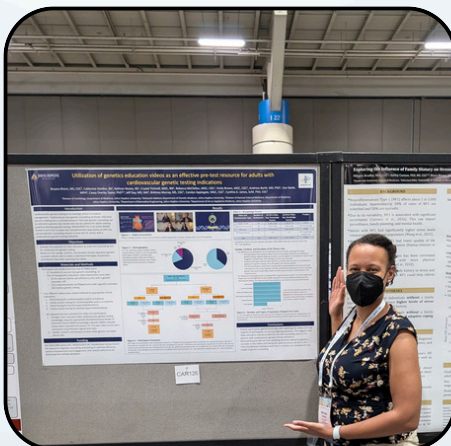
2024 NSGC ANNUAL CONFERENCE - NEW ORLEANS



Rebecca organized a conference session and spoke at the National Society of Genetic Counselors Annual Conference. The session was titled: Not Just a Carrier: Medical and Emotional Impact of Being a Carrier for an X-linked Disorder brought together health professionals and patients to highlight the challenges experienced by women who are heterozygous for X-linked disorders. There are more than 500 known X-linked disorders, including many inherited heart conditions. She is pictured here along with the other speakers in the session.



Bryana submitted an abstract to NSGC and was selected to present a subset of data for our RESEQUENCE-GC research study. Additionally, she volunteered at the cardiovascular genetics special interest group (SIG) booth in the exhibitor hall, passing out resources to all who stopped by.



2025 HEART RHYTHM SOCIETY MEETING - SAN DIEGO

Many of our CIHD team members presented at the annual Heart Rhythm Society meeting in San Diego this year. Pictured from left to right are: Dr. Andreas Barth, Rebecca McClellan, Brittney Murray, Crystal Tichnell, Dr. Cindy James, and Dr. Richard Carrick.



Rebecca (third from the left) was part of a panel at HRS titled What's New in the Implementation of Postmortem Genetic Testing – How Postmortem Genetic Testing is Implemented in the Clinic.



Genetic counselors, Crystal Tichnell (left) and Rebecca McClellan (right) stopped by the exhibit hall to snuggle some puppies in between presentations! 🐾

Additional Talks
 Brittney – Optimizing Genotype-informed Approaches for Arrhythmogenic Cardiomyopathies – Building a Pipeline for Genetic Testing – What is the Optimal Genetics Counseling and Testing Strategy for ACMs
 Cindy – Phenotypic Differences in Genotypic Arrhythmia Cardiomyopathy Subcategories – Exercise Tolerance in ACM Subcategories AND Desmoplakin Cardiomyopathy, Inflammation, and Arrhythmias

2025 MDCGC ANNUAL CONFERENCE - CLARKSVILLE, MD

Lots of our CIHD genetic counselors attended and presented at the Maryland and DC Society of Genetic Counselors Annual Conference. Pictured from left to right are: Dr. Cindy James, Emily Brown, Brittney Murray, Crystal Tichnell, and Bryana Rivers.



Bryana (left), Emily (middle), and Cindy (right) did a joint talk at the 2025 MDCGC conference titled, Novel Service Delivery Models to Improve Access to Genetic Testing



Brittney presenting a talk titled, The Increasing Specialization of Cardiac Genetic Counselors



DNA BANKING AT JOHNS HOPKINS

Dear Patients and Families,

As many of you know, sudden cardiac death is a heartbreaking event that can leave families searching for answers. One crucial tool in uncovering these answers is DNA banking with an option for postmortem genetic testing. Unfortunately, the laboratory we and many other hospitals have relied on for this service is no longer able to offer DNA banking, leaving us scrambling to find a solution.

We are committed to ensuring that families who have lost a loved one to sudden cardiac death still have access to these vital services. To achieve this, we are working to establish our own in-house DNA banking program. However, setting up such a facility requires significant resources and we need your help.

By contributing to our program, you will be supporting:

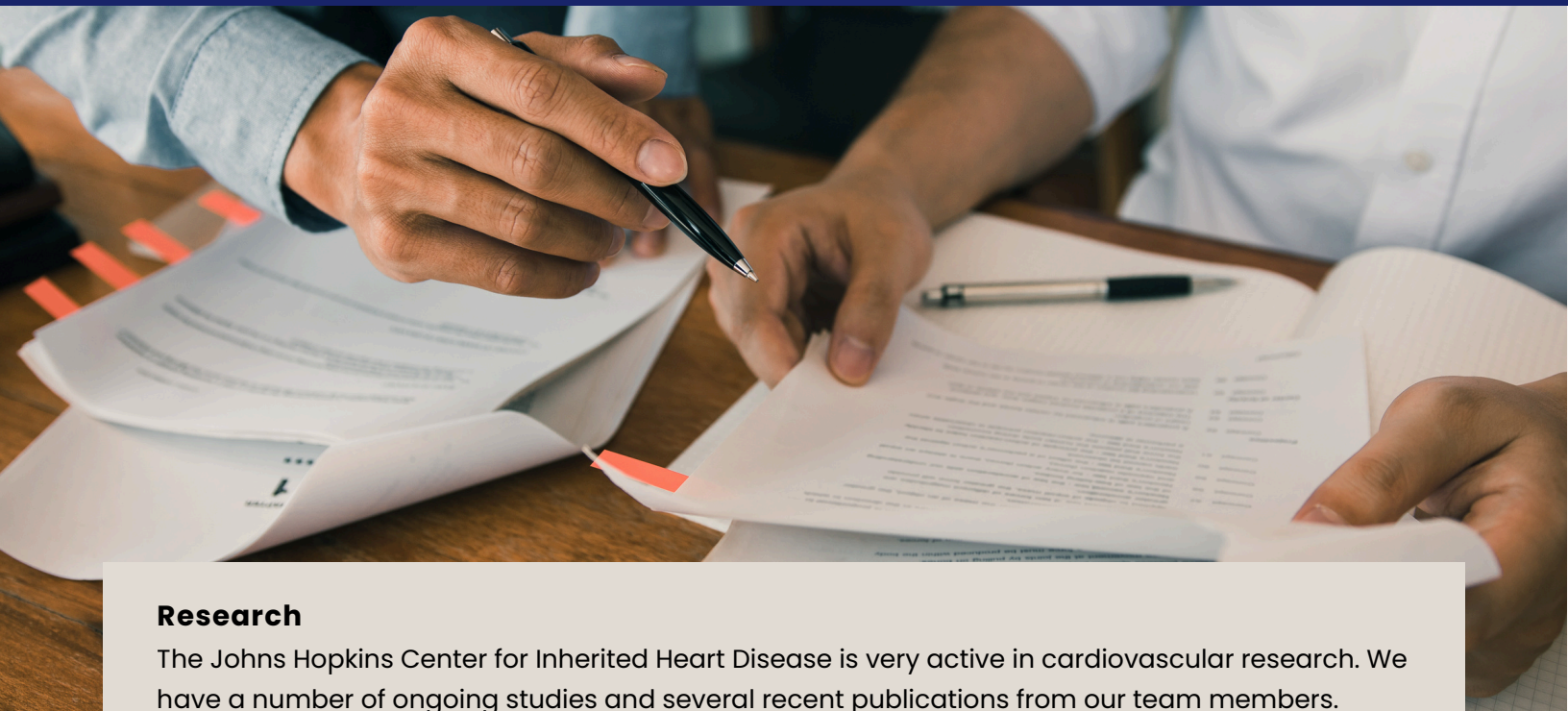
- **The preservation of genetic information:** This is an important resource for surviving family members, for who long-term storage of a loved one's DNA can provide access to postmortem genetic testing both now and in the future as genetic technologies expand.
- **Continued research and better understanding of sudden cardiac death:** Your donation will support ongoing studies aimed at uncovering the genetic causes of these tragic events, potentially leading to better prevention and treatment strategies.
- **Peace of mind for grieving families:** Ensuring they have access to answers during an incredibly difficult time.

Your generosity can make a profound difference. No donation is too small, and every bit helps us get closer to our goal of establishing a reliable, in-house DNA banking service. Together, we can provide hope, support, and answers to families in their time of need.

Thank you for your kindness and support.

Kind regards,

Your team at the Johns Hopkins Center for Inherited Heart Disease



Research

The Johns Hopkins Center for Inherited Heart Disease is very active in cardiovascular research. We have a number of ongoing studies and several recent publications from our team members.

About the CIHD Research Registry

The Johns Hopkins Center for Inherited Heart Disease is dedicated to advancing cardiovascular genetics research as it relates to the health and care of our patients. The first step in getting involved in our research is joining our research registry: "Genetic Investigation of Inherited Cardiac Conditions." The purpose of this study is to learn more about the genetic causes of heart conditions. We also study which gene(s) cause patients with inherited heart conditions to vary in symptoms, outcomes, and rates of disease progression.

What Does Participation Include?

Participation includes giving us permission to review your cardiac medical records. We may invite you to submit a DNA sample. If we do, we will send you a DNA collection kit by mail or collect a sample in clinic. There is no financial compensation for joining the study.

When you join we will also ask you if you are interested in being invited to join other studies specific to your condition or genes in the future.

Who Can Join?

Adults and children with an inherited heart condition and their family members.

How to Join

To learn more, please contact us at GeneticHeartStudy@jh.edu.

Here's How to Donate

Click here:

[DONATE TO SUPPORT THE MISSION OF CIHD](https://secure.jhu.edu/form/heart)



Or copy and paste the following link into your browser:
<https://secure.jhu.edu/form/heart>

Support the Johns Hopkins Heart and Vascular Institute

To contact the Heart and Vascular Institute Development Office:
Lisa Hammann, Director of Development
Fund for Johns Hopkins Medicine

600 North Wolfe Street
Blalock 536
Baltimore, MD 21287

443-287-7384
Hopkinsheart@jhmi.edu

Questions or problems? Please email the [online giving administrator](#).

GIFT INFORMATION

IMPORTANT NOTE: Make sure to select Center for Inherited Heart Disease in the drop down

\$50	\$100	\$250	\$500	Other
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You all are why we do what we do here at the Johns Hopkins Center for Inherited Heart Disease. We appreciate your support and we're excited to continue growing together with you! Let us know what you want to see in the next newsletter! Email us at cardiacgenetics@jhmi.edu!