

Edition:  
Spring  
2018

Cure HHT

# newsletter

News & Information for the HHT Community

## Awareness is the First Step

June is **HHT Awareness Month**, and an opportunity to bring more attention to the disease by educating the public, hosting events, and uniting behind our common cause. On June 23rd the HHT community around the world comes together to celebrate **HHT Global Awareness Day**.

HHT affects 1.4 million people, but 90% are undiagnosed. Raising awareness is key to increasing diagnosis and treatment. Scientists and medical professionals raise awareness of HHT through publications, research and conference presentations. Now's your chance!

**Take Steps to Cure HHT** and help raise awareness through your actions. **Step Out With Us!** at a series of walks and family days in Boston, Chicago, and New York. **Take a Giant Leap Forward** and become a fundraiser for Cure HHT by creating your own event, or joining other events in our 50 State Challenge. You can have national and international impact through social media. Connect and share via Facebook, Twitter and Instagram, and remember to use the hashtag **#TakeStepsToCureHHT**.

Help us **Step Up The Fight** with your generous donation to **Cure HHT** or give as a memorial or tribute. Your gift will impact yourself, your family, and future generations!

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2018



**The  
Year of  
Engagement**



## Message from Marianne

**J**une is HHT Awareness Month and we are making greater strides in getting the word out on HHT to the public, medical professionals, government agencies; yet, there is still much work to be done. Over the last 15 years, we have continued to be contacted each year by hundreds of new individuals, which is an indication there are thousands of families still not aware of a diagnosis that will change their lives.

Increasing HHT visibility is critical! In this issue, you will find a number of ways you can join in the effort to participate in any way you are comfortable. It is through awareness that more people become educated and interested while also attracting more supporters and thereby more resources to our cause.

One example on the federal level was our decision to target our legislative efforts to educate a Senator about HHT who then led the effort to include HHT in the DOD Congressional Medical Research Program. Since 2015, our talented HHT researchers have been awarded \$15,885,000! This successful effort led to new research funding sources that will usher in new therapies in the next few years to benefit our community on our way to a cure. By sharing your story and those of your family with others who may be in a position to help, together we will grow our organization and our ability, bringing game changing therapies to you faster.

### Join us in Stepping Up to participate!

Yours in good health,

Marianne S. Clancy, RDH, MPA  
Executive Director, Cure HHT

**How do you want  
to contribute toward creating  
an HHT-free future?**

Email me any time at  
[marianne.clancy@curehht.org](mailto:marianne.clancy@curehht.org)

### Cure HHT welcomes a new board member to the team

"It is an honor to serve on the Cure HHT board of directors. As a physician who also has HHT, the organization has been an invaluable resource. Now I am looking forward to doing my part. When my daughter heard that I had been elected, she texted, "Proud of you! Cure HHT is doing good for people!" –Gary McKee, MD





## CONNECT WITH US



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[youtube.com/HHTFoundation](https://youtube.com/HHTFoundation)



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<https://curehht.wordpress.com/>

Stay current on HHT clinical trials, education programs and community events. Sign up to receive electronic announcements by registering at [www.curehht.org](http://www.curehht.org) or send us an email at [hhtinfo@curehht.org](mailto:hhtinfo@curehht.org)

### CURE HHT NEWSLETTER

**Marianne Clancy, RDH, MPA**

*Editor*

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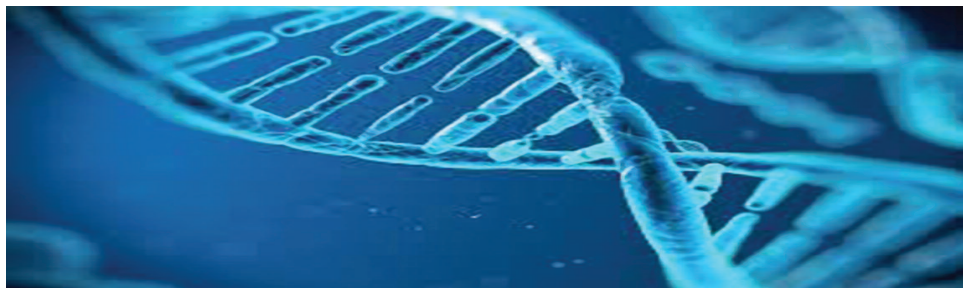
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# Genetic Testing and HHT



To avoid catastrophic medical events, it is important to identify all members of your family, and extended family, that have inherited HHT. Genetic Testing is the only way to diagnose HHT for a person who is at risk for the disease but who does not meet the clinical diagnostic criteria. The first step is to identify the family's HHT genetic alteration (variant) which is accomplished by testing a clinically diagnosed family member. Genetic counseling is a critical component of genetic testing because it allows patients to make an educated decision.

Print the **NEW Genetic Testing and HHT Brochure\*** from our website at <https://curehht.org/resource/genetic-testing-and-hht/> or view scientific literature, articles and webinars on the Resource Library by entering "Genetic" into the search bar.

You can learn more about genetic testing or find an HHT genetic testing lab by visiting our website at:  
<https://curehht.org/understanding-hht/diagnosis-treatment/genetic-testing-hht/>

*\*This brochure was created in collaboration with Cure HHT, Impact Genetics and the genetic counselors at the University of Utah and Yale University HHT Centers of Excellence.*

## Dr. James Gossage Receives Robert E. Berkman Leadership Award



James Gossage, M.D.  
Medical Director, Cure HHT  
Director, Augusta University HHT Center of Excellence

*"Dr. Gossage has been instrumental in the expansion of HHT Centers of Excellence, the development and implementation of the North American Study of Epistaxis, and the creation of the physician forum. He continues to be a compassionate physician and a leader in the HHT community. Dr. Gossage embodies the qualities that Bob Berkman set forth to guide our organization's mission."*

*—Marianne Clancy, Executive Director*

# New HHT Conference Paradigm

**Cure HHT** decided to take our conference on the road in 2018. Instead of hosting one large conference every two years we will be hosting multiple events in areas that are underserved but have HHT Centers of Excellence in neighboring states. This spring we traveled to **Scottsdale, Arizona** and **Nashville, Tennessee**. The smaller, more intimate environment allowed the participants to chat one-on-one with doctors and scientists and engage with other HHT patients during lunch and a special reception. We had over 140 people attend these two events, traveling from as far as Australia, and the feedback has been amazing.

*"These conferences are amazing and such a huge wealth of information.  
Thank you Cure HHT."*

*"I liked the smaller numbers. Easy to meet more people."*

**Mark your Calendars!**

We are coming to the **Ann Arbor, Michigan** on **October 20-21, 2018**.

*Stay engaged with Cure HHT! Make sure we have your most current email and postal address for Save the Date information by emailing [hhtinfo@curehht.org](mailto:hhtinfo@curehht.org).*

## Thank you!

–Silver Sponsors–



**Medtronic**  
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–Bronze Sponsors –



**impact genetics**  
advancing genetic diagnostics

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–HHT Center of Excellence Speakers –

Augusta University, Cincinnati Children's Hospital, Johns Hopkins, University of Arkansas for Medical Sciences, UC Irvine, UCLA, UCSD, University of Colorado, University of Toronto and University of Utah with special thanks to Barrow Neurological Institute, Mayo Clinic, Impact Genetics and Keith Taylor for their contributions to the program.

**Give Aways** – BleedCEASE, BON-NARE, NoseBud

**Scholarship Fund Donors** – Their support allowed 4 adults to attend these educational events.

And the many **volunteers** who assisted us with registration.



#TakeStepsToCureHHT  
#HHTAwareness2018

The more steps we take to build awareness of HHT, the faster we will find a cure! Here is your biggest opportunity this year to rally and raise awareness with friends and family members around the world as a unified voice!

## THE FIRST STEP IS AWARENESS - JUNE IS HHT AWARENESS MONTH

HHT affects 1.4 million people across the world, but 90% of those people are undiagnosed. Raising awareness for this disease is key to increasing diagnosis and treatment for all. And, raising awareness of HHT allows for more doctors, nurses and medical professionals to learn about the different ways to help HHT patients.

### Here are some ways to help build awareness of HHT:

- An HHT diagnosis means there is a whole family of potentially affected people. Download this illustrated summary to learn more: <https://curehht.org/resource/diagnosis-misdiagnosis-hht/>  
Take the time to have a family conversation about HHT today!
- Talk to your health care providers about HHT
- Write a letter to the editor of your local paper
- Share Your Story! We've made it easy for you with an online form: <https://curehht.org/share-your-story-form/>
- Be a Blogger! Visit our blog at <https://curehht.org/blog/> for information on how you can contribute

## 2018 IS THE YEAR OF ENGAGEMENT - STEP OUT WITH US!

Our HHT community may be small, but we are mighty. Cure HHT events focus on raising awareness, educating the public and empowering all to act on behalf of people with this disease. Check out all of our upcoming events here: <https://curehht.org/get-involved/upcoming-events/>

### Step out with us! Here are a few different events to get involved in:

- HHT Walks and Family Days
- HHT Fundraising Events
- HHT Patient Conferences!



## TAKE A GIANT LEAP FORWARD - FUNDRAISE FOR A CURE!

If you are willing to venture further, become a fundraiser for Cure HHT and help us make big strides towards finding a cure. We have a number of different opportunities, and you won't have to go it alone. We'll help guide you step-by-step and, where possible, team you up with other community members who are also looking to make a difference! Get more details here: <https://curehht.org/get-involved/fundraise-for-a-cure/>

### Here are some ways to help build awareness of HHT:

- 50 State Challenge - People across the country are stepping up to the Challenge! Start your own event or join a group!
- Cure HHT Champions - Raise money while training for the athletic event of your choice - running, biking, weight lifting -- you name it!
- Facebook Fundraisers - For a birthday, a memorial or a tribute. It's easy and it works!
- Create Your Own Fundraiser - The sky's the limit - let your imagination soar! We are here to help with your idea.

## IMPACT GLOBALLY - JUNE 23<sup>rd</sup> IS HHT GLOBAL AWARENESS DAY

Through social media, you can connect with and have an impact on people all over the world. Watch for more information about **Global Awareness Day** coming soon!



Visit our Awareness page at <https://curehht.org/get-involved/raise-awareness/>

### Here are some ways to raise awareness through social media:

- Connect and Share on Facebook, Twitter, and Instagram
- Invite friends and family to "like" the Cure HHT Facebook page
- Use the hashtags #TakeStepsToCureHHT and #HHTAwareness2018

## NOW, WE'RE ASKING YOU TO STEP UP THE FIGHT!

We know that not everyone can create a fundraising event or do a walk-a-thon, but we are asking you to participate in any way you are comfortable, and help make this Awareness month the best ever for Cure HHT.

**Great news!** Your gift before the close of our fiscal year on June 30th will be **matched dollar for dollar up to \$50,000**. Double your impact today!

Use the enclosed envelope to make a generous tax-deductible donation, or log on to: <https://curehht.org/TakeSteps> **Our sincere thanks for all that you do!**

# The JOURNAL of PEDIATRICS

## Applicability of the Curaçao Criteria for the Diagnosis of Hereditary Hemorrhagic Telangiectasia in the Pediatric Population

*Kristy Pahl, MD, Arkopal Choudhury, MS, Katie Wusik, LGC,  
Adrienne Hammill, MD, PhD, Andrew White, MD, Katharine Henderson, MS,  
Jeffrey Pollak, MD, and Raj Kasthuri, MD  
(Journal of Pediatrics, 2018)*

Diagnosis of HHT in children can be challenging, as they often lack nosebleeds and telangiectasia, the telltale symptoms of HHT that most patients and physicians are trained to look for. Since children with HHT may not demonstrate these symptoms, it is uncertain whether the Curaçao clinical diagnostic criteria is appropriate to use in children. These criteria have been validated in adults with HHT. This group of clinicians conducted a multicenter study to evaluate the accuracy of the Curaçao criteria for the diagnosis of HHT in those 21 years of age and under. The study included almost 300 children evaluated at the HHT centers at UNC Chapel Hill, Washington University St. Louis, Cincinnati Children's Hospital, and Yale University.

This study demonstrated that the Curaçao criteria are not accurate for the diagnosis of HHT in those under the age of 21 who meet 2 or fewer criteria. Genetic testing is therefore preferred in this scenario to diagnose or exclude HHT. The criteria were accurate in diagnosing HHT in those who meet 3 or 4 criteria; in these cases, genetic testing is deemed to be unnecessary. Additionally, this study showed that while nosebleeds and telangiectasia develop with increasing age, children develop AVMs at the same rate as adults. This highlights the importance of early diagnosis of HHT so that appropriate AVM screening can be pursued. All at-risk children should be evaluated in childhood for HHT as early detection of AVMs allows for appropriate treatment before complications occur.

**Share this important article with your child's pediatrician and family members who are at risk without a diagnosis.**

*FULL article is available in the Cure HHT Resource Library at:*

<https://curehht.org/resource/applicability-curaçao-criteria-diagnosis-hht-pediatric-population/>



# What's New in HHT Research?

Three years ago with your support, Cure HHT funded the Young Scholar program. These young researchers have completed their work and continue their HHT research. We will highlight one project per newsletter to demonstrate the impact of their work.

## Role of Endoglin Mutations in HHT and High Output Heart Failure



Simon Tual-Chalot, PhD,  
Newcastle University on  
Tyne, United Kingdom

High output heart failure occurs when the heart is working overtime to supply the body with blood. This is thought to happen in HHT patients who have severe anemia or multiple arteriovenous malformations (AVMs) in the liver. Anemia occurs when the blood contains too few red blood cells that are responsible for carrying oxygen to the organs and tissues. To compensate, the heart pumps more to circulate the blood cells and oxygen. AVMs are abnormal connections between an artery and a vein and bypass a capillary system that normally is responsible for delivering oxygen-rich blood to tissues. When specific organs, like the liver, have multiple AVMs, not enough oxygen is getting to the organ and the heart must work harder to supply it. AVMs and their small counterparts, telangiectasias have the tendency to bleed and cause anemia. Over time, the heart gains size to help it pump more blood, but it cannot work hard enough to keep up with the oxygen demand of the body.

HHT1 patients have a mutation in endoglin, which is a protein on the surface of the cells that line the blood vessels called endothelial cells. Using mice, Dr. Tual-Chalot and his team previously investigated the role of endoglin in early development and how it causes AVMs. Currently, they are trying to understand endoglin's role in the development of AVMs in adult life, as most HHT patients form AVMs in adulthood. Those with severe AVMs also develop high output heart failure, presenting the need to understand the connection between endoglin and heart failure.

In their current work, they hope to learn more about how changes in the endoglin protein change the function of the heart. Studying this can reveal when and how badly the heart goes into high output heart failure. This is important to know because it can give medical providers clues on how to treat their patients or intervene before the heart goes into heart failure.

In Dr. Tual-Chalot's research, it was found that loss of endoglin from endothelial cells causes AVMs through rapid cell growth and changes in the smooth muscle in the blood vessels. Mice without functioning endothelial endoglin were given something to stop cell growth (inhibitor of vascular endothelial growth factor's receptor 2), and they were protected against high output heart failure.

The researchers have a mouse model without functioning endoglin and no AVMs, but high output heart failure is still present. This suggests high output heart failure can occur in the absence of AVMs, which was not previously thought. They are now trying to investigate what other vascular factors are present in HHT patients with endoglin mutations that may be causing heart failure.

*Special thanks to Cassi Friday who assisted with the scientific summary.*

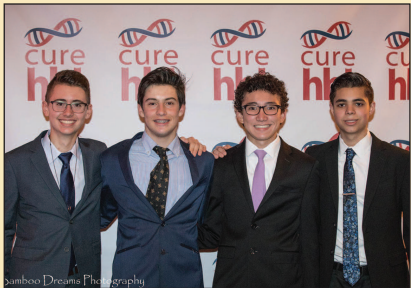
# 6th Annual Night of Hope Event: A Night to Remember

Over 200 people spent this past Cinco de Mayo a little differently in Livonia, MI. They went in style, donning their dancing shoes to support Cure HHT at the 6th Annual Night of Hope, a black-tie event hosted by Jody and Clay Nissan.

The guests had their cake auction – and ate it too! Aside from the games, prizes, auctions, stories and live band, the generosity of those attending this event over the past six years has raised \$300K + for Cure HHT Resarch Programs.

It was an evening full of wonder, amazement and remembrance. Above all, it was a night of hope for the HHT community across the globe.

A special thank you to the Nissan family and their friends who volunteered countless hours that lead up to this event. Without them, it would not have been possible!



We would also like to recognize and thank the  
6th Annual Night of Hope sponsors:



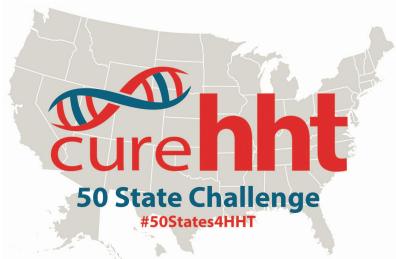
# 50 State Challenge Updates & News

We launched something special at the start of 2018 and as we are almost halfway through this year, our **50 State Challenge** is in full swing!

Our Challengers hail from places like Hawaii, Oregon, Texas and Massachusetts, and they are cooking, running, writing, biking, stretching, Facebook fundraising and everything in between to help us change the future of HHT this year.

Our goal is to have every state in the U.S.A. represented by this December. We challenge you to help us make this goal possible. There has never been a better time to join us and take on the 50 State Challenge!

To learn more and to join us, visit the **Cure HHT 50 State Challenge** website today!  
[www.curehht.org/50-state-challenge](http://www.curehht.org/50-state-challenge)



**New! A Map of Our Challengers!**

*You can now see who has joined our **50 State Challenge** by checking out our map! Head to the Cure HHT 50 State Challenge website to learn more about who is a 50 State Challenger and where our Challengers live!*

**#50States4HHT**

## Join us this June for our HHT Walks and Family Days

We have three HHT Walks and Family Days in three new cities with three HHT Centers of Excellence and all three events are **FREE!**

Join the HHT community during HHT Awareness Month in **Chicago, New York City or Boston!**



**Chicago: Sunday, June 3rd at 10:30 AM**

**Sunset Woods Park, Highland Park**

With HHT Center Director, Dr. Issam Awad, and the University of Chicago HHT Center Staff



**New York City: Saturday, June 9th at 12:30 PM**

**Shore Road and Parkway, Brooklyn**

With HHT Center Director, Dr. Sergei Sobolevsky, and the New York Presbyterian-Columbia University HHT Center Staff



**Boston: Sunday, June 10th at 1 PM**

**Pinebank Promontory, Jamaica Pond**

With HHT Center Directors, Drs. Josanna Rodriguez-Lopez and Raymond Liu, and the MGH HHT Center Staff

### ***Need more information?***

All details about the upcoming HHT Walks and Family Days can be found here:  
<http://curehht.org/get-involved/upcoming-events/>

Want Cure HHT to come to your city for a HHT Walk and Family Day?

Contact us at [events@curehht.org](mailto:events@curehht.org)



**Cure HHT**

PO Box 329

Monkton, MD 21111



***Coming soon: an iOS app  
for HHT patients***

**Everything you need to track your  
nosebleeds, test results, appointments,  
HHT doctors and more! Upload images,  
record your nosebleed frequency and  
severity and send critical information  
to your doctor direct from the app.**

**The app will be FREE –more news soon!**

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