

Edition:
Spring
2017

Cure HHT newsletter

News & Information for the HHT Community

Cure HHT is Here for You

Every day, **Cure HHT** receives calls and emails from our HHT community – people who were just diagnosed, aren't sure where to go for treatment, are wondering how to get their children screened or are fearful of the disease. And every day, for the past 25 years, we've been here to help because we truly care.

Cure HHT is more than an organization. We are a cause – your cause – and with your support we will continue to be here for you another 25 years or until a cure is found.

Your generosity is what fuels our important work of recruiting young researchers, building a community of HHT doctors, providing you the most current HHT information and bringing you together with fellow HHT families.

You are an integral part of **Cure HHT**. Your support helps us make the world a better place for you.

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Message from Marianne

Our organization serves you, the Cure HHT community, as the only organization solely devoted to the welfare of HHT patients and families. We take this responsibility seriously and have recently adopted four goals to achieve our mission:

- **FIND** HHT patients by increasing awareness of HHT.
- **TREAT** the symptoms with advanced therapies.
- **CURE** the disorder through medical or genetic therapies.
- **FUND** the goals from public and private sources.

We plan to do this by continuing as the central hub for your support and resources, organizing the global agenda for all parties with an interest in HHT and sparking advancements in the medical, scientific and clinical communities.

We have limited resources and rely on your help to achieve these goals together!

“Find” Initiatives

Identifying HHT patients and welcoming them to our community improves the lives of patients and families. Our priority is to expand awareness and education by improving your access to timely and up-to-date information. Cure HHT strives to be the primary global educational resource for accurate information.

“Treat” Initiatives

Cure HHT has been instrumental in significant HHT treatment advancements, and will continue to act as a catalyst for improved care. We plan to update screening and treatment recommendations for children and adults while exploring new HHT Center models to ensure standardized HHT care across the US.

“Cure” Initiatives

Our highest calling is to foster the development of lasting cures for the underlying mechanisms and causes of HHT. We will pursue this goal by intelligently investing in and recruiting the best HHT research while advancing our own critical programs like the HHT Outcomes Registry.

But we need your help to make this happen!

I consider this message to you the most important calling within our 25-year-old organization! The time to act is now. Let's create the future we want for our families - a long, healthy and fruitful life. Please join us in making this a reality!

Your partner in finding a cure,

Marianne S. Clancy, RDH, MPA
Executive Director



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youtube.com/HHTFoundation



instagram.com/cure_hht/



<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 or send us an email at hhtinfo@curehht.org.

CURE HHT NEWSLETTER

Marianne Clancy, RDH, MPA

Editor

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Empowerment by the Numbers

Though living with HHT can come with questions and uncertainty, **Cure HHT** is committed to being there for you every step of the way. You want answers and we want to share them with you, whether that be on the phone, through email, in factsheets, at events or on Facebook. We want to make sure you have all the information needed to empower yourself and your family.

Take a look at the ways we were able to help our HHT community this past year!

Provided **555** center referrals



Responded to **540** email questions

Answered **1,042** calls with HHT inquiries



Educated **1,122** webinar attendees

Informed **5,034** people with
Cure HHT eNews updates



Empowered the HHT community
with **23** awareness events

Connected **6,491** people on
social media with HHT information



Quick Facts for Genetic Testing

Genetic testing is a crucial part of being diagnosed with HHT. Not only can it guarantee you receive the correct treatment, but it makes testing cheaper and easier for other family members once your HHT gene is identified.

Here are some quick facts about the process:

What are the known HHT genes?

- ENG (HHT1)
- ACVRL1 (HHT2)
- SMAD4 (affects 2% of people with HHT)
- BMP9/GDF2 (affects <<1% of people with HHT)
- Unknown mutation (affects 3% of people with HHT)

Why should you get a genetic test?

1. To identify your family's HHT-causing mutation and the HHT genetic subtype.
2. To test if an "at-risk" family member¹ has HHT using your family specific gene.
3. To confirm a HHT diagnosis in someone who may have HHT, but does not meet clinical diagnostic criteria based on observable symptoms alone.

¹ *If an HHT mutation has already been identified in a family, genetic testing of at-risk family members for this known mutation is recommended. Unless HHT is ruled out by genetic testing, all children of a parent with HHT should be screened for brain and lung AVMs.*

What are the possible results of the test after my family's gene has been identified?

1. **Positive** for a "pathogenic" (HHT-causing) mutation. A positive result would make it possible to test other people in the family for this precise mutation in order to determine whether or not they have HHT.
2. **Negative**, meaning the lab didn't find a mutation in one of the HHT genes. This could mean the person tested might not have HHT or has an HHT-causing mutation that cannot currently be detected in the genetics laboratory.
3. **Variant of Uncertain Significance** is detected in one of the HHT genes, but it is not possible to predict with certainty if it is the cause of HHT.

To learn more, visit <http://curehht.org/living-with-hht/genetic-testing/>

5 Cure HHT Grants Fund Research Breakthroughs

Research is the answer to curing HHT, and it is Cure HHT's highest priority. Our Young Researchers program attracts the best and brightest to HHT research early in their careers, resulting in continued dedication to the cause. Preventing this disease in the next generation means securing breakthroughs now.

Cure HHT was able to award \$30,000 in grants to the following young scholars with your support. Here is what they have been up to over the last year!



Hongyu Tian, PhD with Duke University

Goal: To determine endoglin's role in regulating the biology of vascular smooth muscled cells, the cells responsible for **stabilization of blood vessels**, during developmental angiogenesis and its relation to HHT.

Update: Using CRISPR technology, the study found important new information about the weakening of blood vessels during development.

Next Steps: The study will expand into mice models and the same steps will then be repeated using HHT cells.



Whitney Wooderchak-Donahue, PhD with University of Utah and ARUP Laboratories

Goal: To identify additional genetic modifiers in HHT to determine if the variability of HHT is caused by mutations in additional genes or **genetic modifiers** critical to vascular development.

Update: More than 160 samples have been collected from two families with ACVRL1 mutation spanning eight generations, including relatives as distant as 13th degree.

The access to distant relatives' RNA facilitates identification of genetic modifiers that may influence development of AVMs in certain family members.

Next Steps: Select patients will undergo transcriptome sequencing, and data will be compared against controls to identify genetic modifiers of AVMs in HHT. This data will be used to guide subsequent experiments in which RNA will be evaluated and analyzed to identify new genes and genetic modifiers.



Simon Tual-Chalot, PhD with Newcastle University

Goal: To investigate how endothelial endoglin maintains adult vasculature to protect against **high output heart failure** in mice models as it is associated with HHT.

Update: The study provided new, important discoveries about how mechanism procedures affect heart failure in HHT.

Next Steps: Understanding the relationship between endoglin function and VEGF signaling will be a major focus moving forward.



Wan Zhu, PhD with the University of California, San Francisco

Goal: To treat **brain AVMs** in HHT patients through gene therapy using a noninvasive strategy injecting a viral vector into the localized site to bind VEGF and inhibit VEGF pathogenic effects, and to extend this treatment for AVMs in other organs.

Update: The findings of this study have been submitted for publication in the Journal of Stroke. Once the findings are publically

presented, more details will be made available.

Next Steps: Testing will continue in mice models to develop a ground-breaking treatment.



Chadwick Davis, PhD with University of Utah and Recursion Pharmaceuticals

Goal: To discover a **known drug** to be repurposed to successfully treat HHT by characterizing ENG, ACVRL1 and SMAD4 deficient cell lines using RNAi and identifying known drugs that fully or partially ameliorate the HHT phenoprint.

Update: After analyzing 700 quantified features, a screenable phenotype associated with SMAD4 was identified.

Next Steps: Recursion will perform a drug screen of their entire compound library to search for additional targets in ENG and ACVRL1.

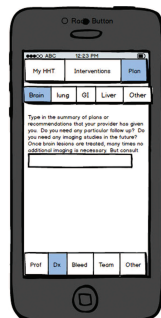
Please note: A more detailed report will be made available once each researcher presents or publishes their findings. The good news is four of them will be presenting in June at the **HHT International Scientific Conference!**

What to Expect from the Year of Possibilities

You have a lot of expectations for the Year of Possibilities, and **Cure HHT** is determined to meet them! These 2017 projects are designed to improve your access to treatment, research and information.

HHT: There's an App for That

Cure HHT is working hard with Dr. Jeffrey Terrell, University of Michigan, over the next year to bring you an HHT app. Patients will be able to track medical treatments, test results, appointment schedules and nosebleed frequency as well as get personalized notifications and reminders to help manage HHT. The app will also provide easy access to Cure HHT educational information on mobile devices. To learn more, watch the HHT app webinar at <http://curehht.org/resources/webinars/>.

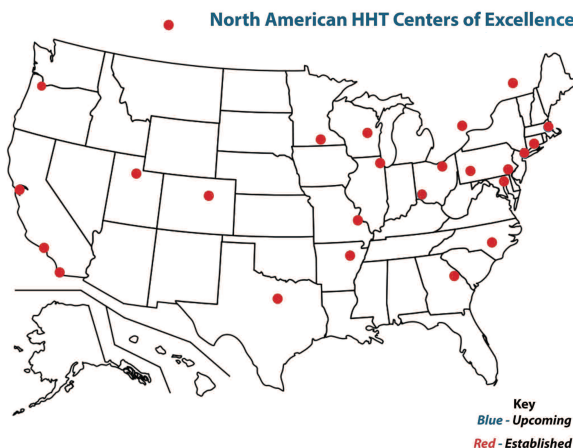


CureHHT.org

A lot has happened since **Cure HHT** launched its website in 2014 – more information is available about HHT than ever before, research breakthroughs are happening every year and awareness continues to snowball. We want our website to reflect all that amazing growth while providing an easy-to-navigate, one-stop-shop experience for you. By the end of the year, CureHHT.org will undergo a major transformation that puts the most important HHT information only a few clicks away, whether you are a doctor, researcher, patient or caregiver.

HHT Center of Excellence Certification Program

Cure HHT made the commitment in 2008 to open two HHT Centers of Excellence a year, and now you have access to 25 centers across North America. But we're only getting started! More than 10 HHT physicians have teamed up with Cure HHT to revitalize the HHT center certification program over the next year. Each center will be required to provide a specific core treatment team and work with a Cure HHT liaison to encourage stronger collaboration on HHT research, education and advocacy. There are already 12 hospitals interested in being part of this new program once finalized!



Join the Cure HHT Champions!

Calling all runners, joggers, walkers, swimmers, cyclists and volunteers!

Find a Cure HHT Partner Race:

Baltimore, MD | Super Run 5K | April 22

Nashville, TN | Rock 'N' Roll Race | April 29

San Francisco, CA | Bay to Breakers | May 21

Denver, CO | Colfax Marathon | May 21

Don't see a team in your area? Create your own today!

Email events@curehht.org

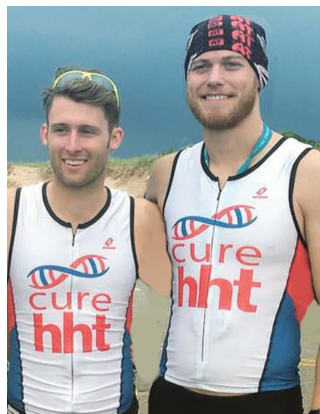
about the endurance event you want to participate in.



Linda DeVizia raised \$12,000 in honor of her son by hosting a fitness class and Cut a Thon at Gerber Salon over the weekend of her 50th birthday.



The Annual Night of Hope welcomed more than 150 people from the Detroit area, raising awareness and more than \$30,000 toward the Cure HHT mission.



Anthony Anzell and Jordan Campbell are swimming 19 miles on August 19 for Cure HHT. Details about attending the Two Crazy Guys Swim for HHT event can be found at: <http://bit.ly/2nZ7bvr>

Upcoming Events

HHT Dallas Walk with Your Doc & Family Day | May 6 | Rockwall, TX

Savannah Walk for HHT | June 3 | Savannah, GA

Stoner Charity Golf Open | June 4 | Kunkletown, PA

See Spot Run Concert | July 1 | Lexington, MI

Cure HHT 5k Run/Walk | August 26 | Ridge, NY

Coming Soon

New York, D.C., Arkansas and Minnesota

A full list of events can be found at

<http://curehht.org/get-involved/cure-hht-team/>

Volunteer Seeks to Save Lives with **Cure HHT Blood Drive**



It's a very real possibility that Cassi Friday's husband, Anthony, and her 8-month-old daughter, Eleanor, may one day need blood transfusions to combat HHT blood loss. Both have been diagnosed with HHT type 1 and have a family history of transfusion dependency.

To encourage blood donations, which may one day treat her family and already saves the lives of HHT patients every day, Cassi held a blood drive this past March in tribute to **Cure HHT**.

"There have been several times when my husband has bled so much, I thought we would have to get an emergency transfusion," Cassi said. "I am the same blood type as my husband and daughter, so I thought giving blood would be a great start."

Cassi contacted the local Red Cross about getting involved and was invited to be part of a drive already scheduled in her area. Not only did the Friday family attend the event, but they were the faces of it, sharing their story in the event press release and marketing.

"We try to donate to **Cure HHT** when we have money, but I feel called to be a more active volunteer," Cassi said. "I'd like to coordinate a blood drive each year to raise HHT awareness."

Cassi's Blood Drive Checklist

- ✓ **Contact your local Red Cross for resources and to set a blood donation goal.**
- ✓ **Find an easily accessible venue and coordinate available dates with the Red Cross.**
- ✓ **Leave enough time to prepare for the event and recruit as many donors as you can!**
- ✓ **Create committees for recruitment, appointments, day of, etc.**
- ✓ **Advertise your blood drive!**
- ✓ **Start recruiting blood donors and set times for them to give blood at the drive.**
- ✓ **Get all contact info to call or text donors a reminder the week before.**
- ✓ **Confirm the venue, parking and Red Cross involvement for day of logistics.**
- ✓ **Get ready to save lives!**



DUBROVNIK
CROATIA

12th HHT INTERNATIONAL SCIENTIFIC CONFERENCE

June 8-11, 2017 Dubrovnik, Croatia

<http://science.hhtconference.org>

Cure HHT is thrilled to announce this year's scientific conference will be held in Dubrovnik, Croatia to further engage international HHT interests and improve HHT awareness in a region that has no known HHT researchers or clinicians.

Advances in HHT treatment and therapies are impossible without the collaboration of the researchers and clinicians at this event - the largest scientific HHT meeting in the world!

Check out the amazing opportunities the HHT International Scientific Conference provides!

1. **Meeting of the Minds** - Colleagues challenge each other to push out-of-the-box ideas and accelerate project progress.
2. **Finding the Right Path** - This weekend is a sounding board for researchers to rethink the direction of their work based on the successes, failures and feedback of others.
3. **In the Know** - Distinguished basic and clinical scientists dive into deep discussion over results, opinions and discussion that can't yet be published.
4. **Finding Consensus** - Through active dialogue and review of published literature at integral conference workshops, clinicians and researchers revise current thinking on controversial topics.
5. **International Collaboration** - A variety of experts from around the world with passionate interest in HHT come together as a single think-tank to take on the HHT problem.

The conference executive summary and all of the accepted abstracts will be published this fall in *Angiogenesis*.



Look how far we've come!

- 1996 – First scientific conference; Edinburgh, UK; 60 attendees
- 2017 – 12th scientific conference; expect 200 attendees
 - 8 invited speakers
 - 57 oral presentations
 - 125 poster presentations

Cure HHT is able to sponsor this important conference thanks to your generous supporting donations.



Cure HHT
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WHAT THE HHT?

A Voice for the HHT Community



Have you checked out the Cure HHT blog?

It's full of stories from HHT patients and families who want to share experiences with you! Join the conversation at <https://curehht.wordpress.com/> or email us to become a blogger.

If you no longer wish to receive this newsletter please contact hhtinfo@curehht.org or 410-357-9932.