

Edition:
Fall
2016

Cure HHT

newsletter

News & Information for the HHT Community

empowerMENT

This year has been about empowering you, the HHT community, to become advocates, not only for advancing awareness and treatment, but also for yourselves and your families.

Empowerment looks different for everyone, and we've been so impressed with how you've chosen to make an impact. Awareness plays a critical role in efforts to expand education, raise the diagnosis rate and save lives around the world, but **awareness does not grow on its own.**

HHT Awareness Month is a pivotal time for the **Cure HHT** mission and everyone affected by HHT. It gives the organization a solid platform to push for advances, but also gives the community an opportunity to take charge of their own healthcare. All that has been achieved is the result of empowered individuals, like you, making sure their voices are heard.

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Message from Marianne

June is my favorite month because every HHT Awareness Month we get to see the benefits of another year working toward a cure. Your dedication and generosity, along with the efforts of Cure HHT staff and the Centers of Excellence, help to continue realizing our goal: to find, treat and cure HHT.

Though the ultimate goal is to find a cure, creating connections within the community for patients to find support and friendship is also a huge motivator for the Cure HHT organization. I was honored to attend two outstanding regional events this June – one in Texas and one in D.C. – where I watched people who began as strangers end the day with hugs.

It touched me to see HHT families in Texas able to connect with one another and have questions answered by Dr. John Battaile, Director of the HHT Center of Excellence at UT-Southwestern. The enthusiasm of our Texas community made Dallas Day for HHT one of the biggest fundraising events of the year.

I also had the opportunity to meet personally with people at the Cure HHT Team DC Walk, and was moved to see so many of you come together to push for change on Capitol Hill. It's wonderful to see this yearly national event, so critical to increasing awareness and support for HHT legislation on a national level, grow each year.

We are raising the standards of awareness, knowledge and care everywhere in the world. This is no small feat – it takes a strong and dedicated community to achieve what you have.

Empowerment is changing the future of HHT.

In partnership,

Marianne S. Clancy, RDH, MPA
Executive Director



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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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Medical Partnerships Lead to Stronger HHT Awareness

It can be hard to feel heard as a patient with HHT, but when you meet a team of doctors who understand what you need, things begin to fall into place. The same is true for **Cure HHT** – though we are a strong advocating voice, it helps to have a team that backs us up to affect change. Each partnership opens the door to further educate the medical professionals who, in turn, end up on *your* team.



Partner: For more than 30 years **National Organization for Rare Disorders** has been a leader in voicing the needs of the rare disease community, driving policies, advancing medical research and providing patient and family services.

Benefits: NORD hosted HHT patient representatives at the American Medical Student Association meeting and two-day NORD Summit conference. Cure HHT has also been incorporated into outreach programs and Global Rare Disease Day.



Partner: **Genetic Alliance** includes more than 1,200 disease-specific advocacy organizations, as well as thousands of universities, private companies, government agencies and public policy organizations. They apply solutions in health and disease, practice and prevention on the local and global level.

Benefits: Genetic Alliance provided IRB approval for **Cure HHT** to recruit for the NIH Brain AVM Study and provides **Cure HHT** with access to a professional discussion forum, disease info research database and seminars related to the rare disease community.



Partner: **Coalition for Imaging and BioEngineering Research** has more than 180 members, including 99 patient advocacy groups representing the full spectrum of diseases that use imaging for detection, diagnosis and treatment. They educate the public about imaging research and advocate for federal support.

Benefits: **Cure HHT** attended the CIBR Medical Technology Showcase on Capitol Hill to educate the radiology community about HHT. **Cure HHT** has also been featured in the CIBR blog, newsletter and social media feeds to educate professionals on recognizing HHT.



Partner: **Society of Interventional Radiology** is a national organization of physicians, scientists and allied health professionals dedicated to improving public health through disease management and minimally invasive, image-guided therapeutic interventions.

Benefits: This partnership has resulted in the designation of the National HHT Patient & Family Conference for a maximum of 18.5 AMA PRA Category 1 Credits™ in continuing medical education, drawing physicians from outside the HHT community to learn about the disease.



BOSTON MARRIOTT BURLINGTON
One Burlington Mall Road
BURLINGTON, MA 01803

October 28-30, 2016
www.hhtconference.org

Spend Halloween weekend unmasking the symptoms, management and treatment of HHT with **Cure HHT** experts, patients, researchers and supporters.

More than 20 educational sessions and workshops, including special aging, family, millennial and youth tracts, are at your fingertips for an entire weekend.

There is something for everyone – whether you have HHT, have family members with HHT, or want to learn about the treatment and diagnosis of HHT. This is your chance to meet others in the HHT community and discuss the disease with leading medical and scientific professionals.

Adult Member - \$275

Adult Non-Member - \$350

Adult Guest - \$295

Youth - \$75

Physician Member - \$375

Physician Non-Member - \$425

Important!

- **Online Registration Only** at <http://hhtconference.org/conference-registration/>.

- **Members receive a conference discount.** If your membership isn't current, choose "I want to become a member" as your registration type for the dues and conference discount to be automatically calculated.

- **Reserve your hotel room** at <http://bit.ly/29SokhC>.

- Learn more about **Conference Scholarship** at <http://bit.ly/29TNoUt>.

Physicians can receive a maximum of 18.5 AMA PRA Category 1 Credits™ through the Society of Interventional Radiology (accredited by the ACCME) while learning about the treatment and management of HHT.

Get Involved

- Encourage your doctors to attend this CME accredited event.
- Share your talents or ask friends and colleagues to support HHT with photography, videography or event planning skills.
- Solicit your company or local businesses for sponsorship and auction items.
- Collect auction items and be feet on the ground to raise awareness about HHT.
- Organize an event leading up to the conference.
- "Work" at the conference as a greeter, youth chaperone or auction organizer.

Contact:

cathleen.kinnear@curehht.org
or 410-357-9932 to donate an auction item or volunteer.

All donors will receive a tax receipt.

Invite your family, local physicians and other medical professionals to join us on October 28-30, 2016. We look forward to seeing you in Boston!

24th HHT Center of Excellence in North America

 **NewYork-Presbyterian**  **ColumbiaDoctors**



Medical Director: Sophie Chheang, MD

Associate Director: David Sperling, MD

Make an Appointment: 212-305-7094 (Adult) contact Helen Pena-Chacon

212-305-5122 (Pediatric) contact Emilio Arteaga-Solis, MD, PhD

nyp.org

"Two years ago, we made a commitment to New York and New Jersey families that we would work very hard to encourage a team of experienced physicians, with our guidance and a HHT mentor center, to come together as an HHT Center of Excellence," said Executive Director Marianne Clancy. "I am so happy to see this work become a reality. I am confident this superb team, which I met during a site visit, will provide outstanding care. We look forward to working closely together in the years to come."

NewYork-Presbyterian/Columbia University Medical Center is the only facility in New York that specializes in the diagnosis, evaluation and treatment of HHT. The newest HHT Center offers compassionate, family-centered multidisciplinary care, led by Dr. Sophie Chheang, a dynamic and action-oriented physician. *At the NewYork-Presbyterian/Columbia University Medical Center, each patient is treated by HHT-knowledgeable physicians who are compassionate and devoted to maintaining the highest standards of care. Dr. Chheang's team includes a full team of pediatric specialists lead by Dr. Emilio Arteaga-Solis.*

Dr. Chheang is a board certified radiologist at NewYork-Presbyterian and an assistant professor of radiology at Columbia University Medical Center. After graduating from the New York University School of Medicine, she completed her Diagnostic Radiology residency at the University of California,



Los Angeles. Dr. Chheang then finished her Interventional Radiology fellowship at NewYork-Presbyterian/Weill Cornell Medical Center, which included additional training at the Memorial Sloan Kettering Cancer Center. In addition to English, Dr. Chheang speaks Khmer and Spanish. She is Board certified with the American Board of Radiology.

Specialties at NewYork-Presbyterian/ Columbia University Medical Center

Cardiology
Dermatology
Genetics / Genetic Counseling
Gastroenterology
Hematology
Interventional Radiology
Neuro-interventional Surgery
Neurosurgery
Otolaryngology (ENT)
Pediatrics
Pulmonology

Knowledge is Power

Cure HHT has created a factsheet to educate your family and health care providers. HHT “masquerades” as many different conditions and is frequently misdiagnosed.

Diagnosis and misdiagnosis of HHT

Hereditary Hemorrhagic Telangiectasia (HHT), also sometimes referred to as Osler-Weber-Rendu (OWR), is a genetic disorder of the blood vessels affecting approximately 1 in 5,000 people totaling 1.4 million worldwide. While HHT affects specific areas or organs of the body, it's important to understand the potential impact on other aspects of life as well. To ensure the best possible health for you or your loved ones, remember to consider how HHT might impact or cause the following: aging, pregnancy, iron deficiency, genetic testing, anemia.

As HHT symptoms are often mistaken for other conditions, knowing the common misdiagnoses can lead to faster identification and significant improvements in quality of life.



AFFECTED ORGANS

HHT does not affect every individual in the same way. Some people may have symptoms in several different parts of the body, while some may show hardly any symptoms at all.

- 1 BRAIN
- 2 LUNGS
- 3 SPINE
- 4 NOSE
- 5 GASTROINTESTINAL (GI)
- 6 LIVER
- 7 SKIN

1 BRAIN

Up to 23% of people with HHT have arteriovenous malformations (AVM) in the brain, and in most cases can be successfully treated. If left untreated, brain AVMs can be disabling or deadly, and because they often have no symptoms prior to bleeding, HHT physicians in North America recommend brain screening for anyone diagnosed with HHT, even infants and during developmental growth periods until age 21.

Common Misdiagnosis: Aneurysm, Cerebral Hemorrhage, Epilepsy, Abscess, Migraine, Stroke

Treatment: Embolization, Radiation, Surgical Removal

2 LUNGS

40-50% of people with HHT have arteriovenous malformations (AVM) in the lungs. Those with HHT1 (Endoglin) are 5-10 times more likely to have pulmonary AVMs (PAVMs) than people with HHT2 (ACVRL1 or ALK1). PAVMs are at higher risk of rupturing when blood volume in the body increases (i.e. during pregnancy). Until PAVMs are excluded by testing, a person over the age of 10 with known or suspected HHT should take antibiotics before all dental work or medical procedures that could introduce bacteria to the blood stream.

Common Misdiagnosis: Asthma, Alzheimer's Disease, Lung Disease, Tumor, Stroke, Polycythemia, shortness of breath, coughing up blood, Infertility, Patent Foramen Ovale (PFO), Migraines, Brain Abscess, Stroke

Treatment: Endovascular Embolization

3 SPINE

Spinal arteriovenous malformations (AVM) are rare, affecting only about 1% of HHT patients. They can cause back pain and sometimes lead to loss of feeling or mobility in an arm or leg. If untreated, spinal AVMs can lead to further motor or sensory deficit, aneurysm, and high blood pressure.

Common Misdiagnosis: Scoliosis, Stroke

Treatment: Endovascular Embolization, surgery, Radiation Therapy

You can find the full version of “Diagnosis and Misdiagnosis of HHT” online at <http://curehht.org/resources/hht-brochures/> along with 12 other factsheets to answer all your HHT questions.

HHT Awareness Month by the Numbers

This June was all about celebrating **HHT Awareness Month** with events, media campaigns and lobby days designed to strengthen awareness. Millions of people were touched by the **Cure HHT** mission - from the HHT community to government representatives, and from medical professionals to the general public.

A huge thank you goes out to the more than 350 participants and donors who made the June events possible and supported the **#AreYouHHT** campaign. We are so grateful to everyone who has continued to be the driving force behind **Cure HHT**, and to all of you who continue to invest in an HHT-free future.



This year's **Cure HHT Team DC Walk** raised more than \$11,000, bringing people together from across the United States to promote HHT awareness in the nation's capital.

HHT patients and supporters came from as far as Michigan to lobby for the **HHT DATA Act**.



HHT families came from all over Texas to the **Dallas Day for HHT** to meet with **Cure HHT** doctors and staff for the first time.

You raised \$30,800 toward the **Cure HHT** mission!

- More than **150 people** attended the events held in Texas, D.C. and California.
- Almost **500 people** joined the **#AreYouHHT** profile picture campaign on Facebook, Twitter and Instagram.
- The new **Cure HHT PSA** video reached more than **1.5 million people** through online views and live events. Watch it now at <http://bit.ly/29TbW2D>
- A group of 12 HHT patients and supporters met with **7 different representatives** about the **HHT DATA Act** on Capitol Hill Day.



The **Dallas** event raised close to \$12,000 for HHT research and treatment initiatives in its first year.



FlyEvent: The Boock family held a sold out indoor cycling event in California with more than 40 bikers supporting **Cure HHT**.

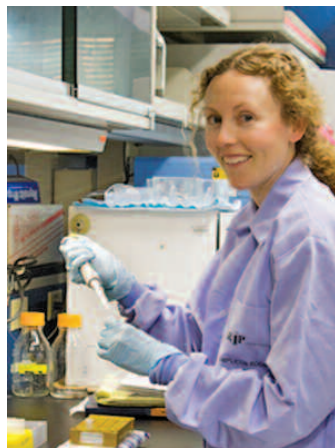
The **Cure HHT PSA** showed twice an hour, every hour at the Indy 500 race over Memorial Day weekend.

One in every 5,000 people has HHT.
#AreYouHHT?

To find out more about hosting your own event, contact us at events@curehht.org or 410-357-9932. To see the list of upcoming events, go to <http://curehht.org/>, click on the Get Involved tab and select Fundraising, Upcoming Events from the sidebar.

Young Researcher Works to Discover New HHT Genes

Whitney Wooderchak-Donahue, PhD, said she couldn't help but jump around her office when she received the \$30,000 **Cure HHT Young Research Scholar Award**. Having studied the molecular genetics of HHT for more than 10 years, it was a dream come true to receive a seed grant to further her research. Almost a year later, Whitney's results are awaiting publication and she's thrilled to use those findings to leverage federal funding to expand the project.



Q: What motivated you to apply for the **Cure HHT Young Research Program?**

A: I really want to find an answer for families who don't know why they have HHT. Research is like a roller coaster - One day you think you've found a new variant that causes HHT and the next you find that same variant in a control sample. We will get there though. I'm determined!

Q: What is the goal of your **Cure HHT project?**

A: To discover additional HHT genes and genetic modifiers using the latest, most advanced genetic technologies. We hypothesize that the clinical variability observed in HHT patients, like multiple AVMs and severe nosebleeds versus no AVMs and few nosebleeds in individuals from the same family, is caused by mutations in additional genes and genetic modifiers critical for vascular development.

Q: What are your next steps?

A: In one year, we have collected about 100 cases with a known ENG or ACVRL1 mutation, many of whom are from two large extended Utah pedigrees with an ACVRL1 mutation. We are currently analyzing the results from our transcriptome experiments and plan to use data from this small study to apply for larger federally funded grants later this year.

Q: Will you continue to focus on HHT moving forward?

A: My future work will focus on understanding the molecular genetics of HHT and other vascular malformation syndromes to improve the understanding and molecular diagnostics of these disorders so more families can have an answer. We all have one goal in mind - to help patients with HHT.

Because of your support, **Cure HHT was able to invest \$150,000 in the 2015 Young Scholar Program, funding five new areas of HHT research.**

YOU can help advance HHT Research!

Did you know you are 10 times more likely to have an AVM brain bleed if you've had a previous BAVM bleed? Did you know a HHT brain AVM involves more high-flow and small lesions than non-HHT brain AVMs?

Well, neither did **Cure HHT** until we began the Brain Vascular Malformation Consortium study. Now more than nine research papers have been published to educate doctors and ensure better treatment for HHT patients. ***That's the power of research!***

Your involvement in **Cure HHT** research is a direct way to impact the future of HHT. ***Get started today.***

Brain AVM Study

This is an ***information gathering study*** to determine what genetic and clinical factors signal high risk for hemorrhage from brain AVMs. This ongoing project helps doctors make better treatment decisions about brain AVMs and drives research in brain AVM therapies. **You don't have to travel anywhere. There's no cost to you.**

Eligibility

- Definite clinical or genetic diagnosis of HHT
- Brain AVM diagnosis, whether it has been treated or not
- Live in the United States or Canada
- Be at least 3 years of age

Bevacizumab (Avastin) for HHT Nosebleeds

Cure HHT is pleased to offer **financial support to offset reasonable travel expenses** to Palo Alto, CA for Bevacizumab (Avastin) clinical trial participants who have a documented financial need. This study is designed to determine if intranasal bevacizumab injection decreases nosebleed frequency and severity compared to a placebo.

Eligibility

- Anyone 18 or older with HHT who would like to have electrocautery for their nasal bleeding AND can complete surveys at the four time points after surgery
- Participant is responsible for travel to Palo Alto, CA and insurance coverage for surgery.

For more information about these and other clinical trials, visit <http://curehht.org/medical-scientific/researchstudies/>, call **Cure HHT** at 410-357-9932 or email research@curehht.org.

Become a Cure HHT Hero at a Super Run Near You!



We've teamed up with organizers behind the **Super Run 5k** and now YOU have a chance to be a **Cure HHT Hero**! You don't have to run or walk (unless you want to!) because we are looking for volunteers to help each event in one of the 20+ locations across the U.S.

To find out more about the **Super Run 5k series**, visit:
www.thesuperrun.com

Instructions:

So how do you join? Contact events@curehht.org to volunteer or participate in the **Super Run 5k** at a location nearest you.

Join Us at a **Super Run** Near You:

COLUMBUS, OH — 9/10/2016
DENVER, CO — 9/17/2016
AKRON, OH — 9/17/2016
LANSING, MI — 9/24/2016
KALAMAZOO, MI — 10/1/2016
INDIANAPOLIS, IN — 10/8/2016
TOLEDO, OH — 10/8/2016
DALLAS, TX — 10/15/2016
SAN JOSE, CA — 10/15/2016
KANSAS CITY, MO — 10/29/2016
ATLANTA, GA — 11/5/2016
JACKSONVILLE, FL — 11/12/2016
FRESNO, CA — 11/20/2016
EL PASO, TX — 12/3/2016
FORT WORTH, TX — 12/10/2016
FORT MYERS, FL — 12/17/2016

*There are future runs in the works for 48 cities, and
with enough interest there can be a **Super Run** in your hometown!*

Street Renaming Memorializes HHT Champion



Qadri and Doreen at the Children's Hospital of Pittsburgh of UPMC as part of the Flashes of Hope campaign.

In 2013, Doreen Blair Skipper began petitioning the New York City Council to rename a street after her beloved son and HHT champion, Qadri Skipper. Her dream was realized on June 21 when she was joined by friends, family, council members and Dennis Routledge, of the Cure HHT Board, to honor Qadri's memory.

"This was not only a testament of his character - helping other pediatric patients with their parent caretakers, the witty and indelible impression he left on others - I wanted to express our gratitude for all who partook in his care," Doreen said of the ceremony which renamed a portion of 110th Street in Queens.

Qadri lost his battle with HHT and Pulmonary Hypertension at the age of 17 in 2012, but not before touching the community around him with his unending optimism, enthusiasm for life and dedication to learning. He's also remembered by students at his high school during a yearly scholarship ceremony, where five awards are given in his memory, and by fellow patients who attribute Qadri as the reason they are still alive.

"Qadri was a special young man who did not allow adversity to define him," Dennis said. "The street naming was the end of a long journey for Doreen, at which she showed her love for her son and could smile again."

Doreen said she will continue to share "**Qadri's Way**" as she moves forward, spreading awareness of HHT and PH to provide the comfort and support that Qadri gave to all he came in contact with.

"I hope when 'Qadri's Way' is Googled, it will shed light and awareness on PH and HHT," Doreen said. "It's not just a nosebleed."



Do you want exclusive access to our educational webinar series? Interested in receiving a quarterly print newsletter with all the latest HHT news? Ready for reduced conference registration fees and VIP reception?

Do you want to be a part of the cure?

Membership is the best way to stay connected to the HHT community and support the 1.4 million people around the world who are affected by HHT.

Join now for only \$55 per year

CureHHT.org

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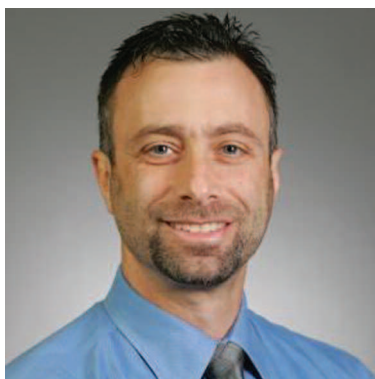
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For More Information, Visit bleedcease.com or Call 1.800.650.6673.

The above is a paid advertisement.

Cure HHT Board Updates

Join us in saying farewell to Scott Olitsky, M.D. and Mike Nolan, two dedicated **Cure HHT** Board Members. We thank them for their years of service guiding the HHT community toward new levels of awareness and a cure.



"When I started on the board in 2009, there were 12 HHT Centers of Excellence and virtually no talk about medications treating underlying causes of HHT. Today, we have 24 centers in the U.S. and have introduced new medications based on research funded by Cure HHT. Although we have a long way to go, awareness is building. It has been a privilege to serve on the Cure HHT Board of Directors."

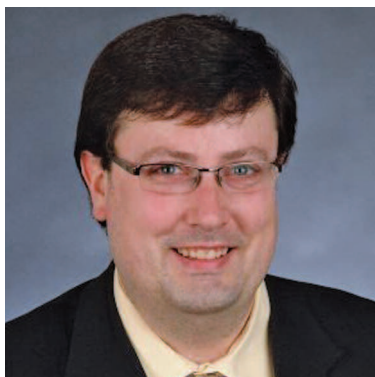
– **Scott Olitsky, M.D.**



"I joined the board in 2002 - our family had suffered great tragedy and I experienced first-hand the lack of screening, treatment and awareness of HHT. We've achieved so much as an organization since in the areas of awareness, treatments, centers of excellence and research. My hope is for HHT to become fully understood, with government funding dedicated to research initiatives and treatment." – **Mike Nolan**

Cure HHT would also like to welcome two new board members to the team!

"While HHT has not affected me or my family directly, I have been touched by the bravery of those that do. I want to make the world a better place and direct my actions at organizations that make a difference, both locally and nationally. I believe Cure HHT is that national influence for good in searching for a cure." – **Ben Holt**



"I'm anxious to better understand the workings of a patient advocacy and research organization, and continue to refine a strategic plan that maximizes the chance of a positive outcome for patients with HHT. I look forward to establishing and maintaining relationships with partners and prioritizing scientific projects to move the organization forward." – **Christopher Gibson, PhD**





Cure HHT
PO Box 329
Monkton, MD 21111

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



You Are Invited!

Location:

Boston Marriott Burlington
One Burlington Mall Road
Burlington, MA 01803
(781) 229-6565

Hot Topics:

HHT 101
Nosebleeds Management
HHT and Aging
Get Personal - HHT in Women
Unraveling Your DNA
AVMs – Tangled Web of HHT
Research and Future Therapies

Continuing Medical Education: (Physicians Only)

Attendees receive a maximum
of **18.5 AMA PRA Category 1
Credits™**. Approved by the
Accreditation Council for
Continuing Medical Education,
provided by the Society of
Interventional Radiology.



October 28-30, 2016

Come with your questions about HHT,
research and treatment!

The weekend includes more than 20 HHT experts
presenting on 25 medical and research topics as well as
a full youth program and empowerment workshops.

Online Registration at www.hhtconference.org.

Invite your doctors to attend for education credit.
It will change how they practice medicine!

Special discount available for Cure HHT members



Cure HHT

PO Box 329

Monkton, MD 21111



**Join the Cure HHT staff, board and community
this October in Boston for the HHT National Patient
and Family Conference.**

Register today: <http://hhtconference.org/>

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