



Edition:  
Spring  
2016

# Cure HHT newsletter

News & Information for the HHT Community

## Join Us in Boston!

Cure HHT has always believed that knowledge is one of the most powerful tools for the HHT community. It is the key, not only to understanding the effects of HHT, but also to spreading awareness and advocating effectively for yourself and your family.

The **National Patient and Family Conference** is a longstanding tradition for Cure HHT. Since the first meeting in 1992 in Ohio, our community has gathered annually to share, learn and grow together. This year the conference will be held in October in Massachusetts, featuring research updates, expert presentations on new and future treatments, empowerment workshops and a special kids program.

Whether you are newly diagnosed or a conference veteran, this meeting is a unique opportunity to expand your HHT education and connect with individuals and families committed to changing the future of HHT. We can't wait to see you there!

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## Message from Marianne

**I**n February, I presented the Cure HHT staff with a challenge – to redefine their individual roles. We pride ourselves on being a cohesive unit, and on our willingness to pitch in wherever needed, but it was time for a little spring cleaning. The goal was to eliminate distractions and focus on the essentials of what we do, and how each of us uniquely serves the mission and goals of our organization.

We have always known that we could ask each other for help, but with the new clarity of our roles and abilities we realized we could also *offer* help. This is what empowerment means to me.

I am asking you to embrace the same challenge I presented to the staff two months ago. On many occasions, I've asked you to consider others affected by HHT. Now, I want you to take some time and think about you. What is your experience? What perspective do you bring to our shared mission? What unique skill, interest or quality do you want to share?

We are enormously fortunate to have a diverse and widespread HHT community. There are many roles to fill, many ways to contribute, many options for changing our future. In this Year of Empowerment, I am asking you to seize the opportunity to pursue the Cure HHT mission in your own way.

What does empowerment look like for you? From spreading awareness, to downloading HHT factsheets, to educating family and friends, to local events or school science projects, there are countless ways to be part of the progress.

HHT affects us all, and it's time for all of us to affect 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](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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# Could Awareness Save Your Life?

Awareness is about more than receiving a diagnosis of HHT, it's about taking steps to educate yourself and those around you about HHT. Knowledge can empower you to act for yourself and your family, but it can also help those who don't know about HHT.

With **HHT Awareness Month** just around the corner, **Cure HHT** wants you to speak up and represent with us. It's time to look within and answer the question, "Are you HHT?" Will you lend your strength to the HHT community as we seek awareness for the 1.4 million people affected by this disease?

**Join Cure HHT in June** for what we hope will be the largest and furthest reaching awareness campaign yet. Stay tuned to [www.curehht.org](http://www.curehht.org) for the launch of **#AreYouHHT**. You could save a life.



"I said, 'What does having bloody noses have to do with my lungs?' That started my true education of Osler-Weber-Rendu Syndrome, or now HHT." **-Mary G., Connecticut**

"I remember the doctor coming in and saying, 'You're a keeper. Your hemoglobin is only four!' That was the first time I had ever heard of hemoglobin, and I didn't have an idea if four was good or bad."

**-Keith S., Ohio**



"I wish I had known about the urgency to be screened. My life has changed forever. I feel fortunate that it was only my sternum and hip, it could have been a lot worse."

**-Anita R., Minnesota**



# HHT Gene Discovery Pushed Advances



**Dr. Marchuk (center) and his 2016 lab team at Duke University.**

Of all the individuals who've helped shape the future of HHT, few have had the impact of Dr. Douglas Marchuk.

After earning his Ph.D. from the University of Chicago, Dr. Marchuk continued his postdoctoral studies at the University of Michigan and it was there he first learned about HHT. With the goal of starting his own genetics lab, Dr. Marchuk consulted with his supervisor, Dr. Francis S. Collins, on a possible HHT project.

Dr. Marchuk then teamed up with Dr. Alan Guttmacher, a medical geneticist who had studied HHT within extended families. Their first breakthrough was mapping the cause of HHT1 to chromosome 9, and then identifying mutations in the endoglin gene, making the researchers the first to identify a specific cause of HHT.

Further research in other HHT families led to the identification of the HHT2 (ALK1) and SMAD4 genes responsible for 90 percent of HHT clinical diagnoses. While experts believe there are yet unidentified genes linked to HHT, the results of Dr. Marchuk's initial project have been a critical foundation for current research, treatments and potential future breakthroughs.

***Cure HHT is fortunate to have Dr. Marchuk as a member of the Global Research and Medical Advisory Board. He is also a Professor of Molecular Genetics and Microbiology and Director of the University Program in Genetics and Genomics at Duke University. Since he first learned of HHT in December 1991, Dr. Marchuk has tirelessly pursued the study of genetics in cardiovascular disease, focusing specifically on diseases involving blood vessels, like HHT.***

## 23rd HHT Center of Excellence in North America

### Massachusetts General Hospital

### Boston, MA



MGH - HARVARD



MASSACHUSETTS  
GENERAL HOSPITAL



**Massachusetts  
Eye and Ear**  
sense life. experience life.



MassGeneral Hospital  
for Children™

Medical Director: Josanna Rodriguez-Lopez, MD

Associate Director: Ray Liu, MD

Patient Services Coordinator: Rosalie Morse

Make an Appointment: 617-724-3705

Cure HHT has brought Massachusetts General Hospital (MGH) together with Mass General Hospital for Children and Massachusetts Eye & Ear Infirmary to form the only facility in the state specializing in the diagnosis, evaluation and treatment of HHT.

This collaboration allows them to provide specialized care across a patient's lifespan. The MGH HHT Center offers compassionate, family-centered multidisciplinary care, led by Dr. Josanna Rodriguez-Lopez, coordinating care for adult and pediatric patients. She is joined by an outstanding group of physicians, including Dr. Raymond Liu as the Associate Director, who share her dedication for the multidisciplinary treatment of this disease.

**Dr. Rodriguez-Lopez** has dedicated her career to treating patients with pulmonary vascular disease. As the Associate Director of the Pulmonary Hypertension and Thromboendarterectomy Program at MGH, she specializes in all aspects of pulmonary vascular disease, with a particular interest in



pulmonary arteriovenous malformations. She has several years of experience managing HHT and is looking forward to growing the HHT program as well as developing a strong partnership with Cure HHT and the HHT community.

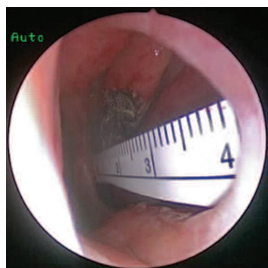
### Specialties at MGH HHT Center

(\*denotes adult and pediatric services)

**Cardiology\***  
**Dermatology**  
**Dentist / Oral Surgeon**  
**Genetics / Genetic Counseling**  
**Gastroenterology\***  
**Hematology\***  
**Interventional Radiology**  
**Neuro-interventional Surgery**  
**Neurology / Stroke**  
**Neurosurgery\***  
**OB / GYN**  
**Otolaryngology (ENT)\***  
**Primary Care (PCP)**  
**Pulmonology\***  
**Radiology**  
**Surgery\***

# Septal Perforation Repair

By Dr. Jason Hamilton, Director of Plastic and Reconstructive Surgery,  
Osborne Head and Neck Institute, Los Angeles, CA



Nasopharyngoscopy demonstrating nasal septum perforation. A ruler has been inserted into the opposite nostril to demonstrate scale.

Typically, HHT nosebleeds are caused by ruptured telangiectases inside the nose, but can be attributed to other, less common issues as well.

One such cause is septal perforation, which occurs when cartilage separating the two nostrils (known as the septum) develops a hole or fissure. This can cause a variety of symptoms, most notably nose bleeding. In HHT patients, septal perforation is usually attributed to nasal trauma from aggressive laser or electric coagulation sometimes used in treating epistaxis.

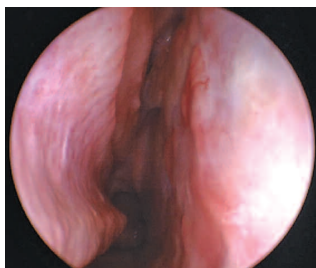
A septal perforation alters the anatomy of the nose, which affects its function. The normal humidity in the

nose is decreased by constant airflow across the weak or torn edges of the perforation, causing the site of the perforation to dry out, leading to a crusted or scabbed area likely to bleed. When left untreated, it causes further deterioration of the condition.

The surgical procedure to repair a septal perforation should be performed soon after a diagnosis, and not as a last resort. The goal is to restore normal nasal anatomy and humidification and to reestablish the structural integrity of the nose – it is not a cure for HHT-related epistaxis.

Surgery typically requires taking healthy mucosa from in and around the nose to construct tissue flaps to repair the damage inside. In HHT patients, any telangiectases around the site of the perforation are ablated during the surgery. Patients generally take seven to 10 days to recover post-surgery, although a protective splint is worn inside the nose for three to four weeks.

This is a surgery that should only be performed by a surgeon with a proven track record for success, which means many people seeking treatment may have to travel out of state to receive expert care. That said, there are many benefits to surgical repair for septal perforation. It has been shown to improve the incidence of nosebleeds, decreasing the need for transfusions and preventing the need for Young's Procedure. Overall, patients frequently enjoy an increased quality of life.



Nasopharyngoscopy of the same patient demonstrating nasal septum perforation repair.

*Dr. Jason Hamilton contributed the medical information for this article. He runs a dedicated team that performs multiple septal perforation repairs each week. If you are considering surgical treatment for this condition, we encourage you to contact Dr. Hamilton or Cure HHT for guidance in pursuing the best possible care.*



## ***Empowering the HHT Community*** ***The 18th National Patient & Family Conference***

**OCTOBER 28-30, 2016 BOSTON MARRIOTT BURLINGTON, MA**  
**[www.hhtconference.org](http://www.hhtconference.org)**

Educating yourself about this disease **empowers** us all to create a future that allows HHT patients to live a normal life, filled with spontaneity and joy.

### **Program**

We are excited to offer a continuing medical education accredited program for health professionals, highlighting two tracks on Friday – Family Planning and Pediatrics or Advanced Stages of HHT – along with robust presentations over the weekend covering PH, anemia, nosebleed management and much more. HHT researchers will share how science is advancing a cure for HHT through drug therapies, personalized medicine and research poster presentations.

The Youth and Teen Program is a great activity for children ages 6 to 16 to take part in age-appropriate fun discussions with HHT specialists to foster better understanding of family history, how to care for themselves and to create bonds with other children affected by HHT.

### **Volunteers**

YOU have the power to change the future of this disease by empowering others at this conference. YOU have the skills, resources and opportunity to spread awareness about this disease.

Sharing your time and talents as a volunteer is a great way to strengthen your community, connect with others and transform your own life. **WE NEED LOTS OF HELP** on a local level.

- Share your talents, or get your talented friends/colleagues to support HHT!
  - Photography
  - Event Planning
  - Videography
- Solicit 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
- Encourage your doctors to attend the CME accredited conference
- “Work” at the conference as a greeter, youth chaperone or auction organizer

### **Have Something Fabulous to Donate?**

- Vacation Stay
- Golf Course Packages
- Sports Memorabilia
- Theme Park Vouchers
- Designer Items
- Autographed Books
- Event Tickets
- Jewelry

Contact:

**[catheleen.kinnear@curehht.org](mailto:catheleen.kinnear@curehht.org)**

or 410-357-9932 to donate an auction item or volunteer.

All donors will receive a tax receipt.





## **BOSTON MARRIOTT BURLINGTON** **One Burlington Mall Road** **BURLINGTON, MA**

**October 28-30, 2016**  
[www.hhtconference.org](http://www.hhtconference.org)

### **Sponsors**

**Cure HHT** offers various options to fit any budget and will customize sponsorship packages. Visit [www.hhtconference.org](http://www.hhtconference.org) to view the Sponsor and Exhibitor Brochure and Agreement.

- Which of our sponsorship opportunities aligns with your company's mission? Or ask about a volunteer giving program?
- Encourage small businesses to become a sponsor or donate an auction item to show support for the HHT community.

Contact [conference@curehht.org](mailto:conference@curehht.org) or 410-357-9932 ext. 101 about conference sponsorship, employer giving/matching programs or employer volunteer programs.

***Partner with Cure HHT***  
***as we continue to change***  
***the future of this disease!***

Learn more about the conference and reserve your hotel room now at [www.hhtconference.org](http://www.hhtconference.org). Online registration will be available in May.

**Cure HHT** members will receive a significant discount on conference registration. Join or renew your membership now at [www.curehht.org](http://www.curehht.org).

### **Thursday, October 27**

4 pm Registration Open  
7 pm Member Reception

### **Friday, October 28**

7:30 am Registration Open  
8:30 am Welcome  
9 am Morning Lectures with Q&A  
12:15 pm Lunch  
1:15 pm Track 1 – Family Planning and Pediatrics  
Track 2 – Advanced Stages of HHT  
3 pm Breakout Sessions  
5 pm Research Poster Session

### **Saturday, October 29**

7:30 am Breakfast  
8:30 am Morning Lectures with Q&A  
12:30 pm Lunch  
1:15 pm Afternoon Lectures with Q&A  
2:55 pm Breakout Sessions  
4 pm Relax  
6:30 pm Research Poster Session / Cocktail Reception  
7:15 pm Dinner

### **Sunday, October 30**

7:30 am Breakfast  
8:30 am Empowerment Workshops  
9:30 am Morning Lectures with Q&A  
12:15 pm Closing Remarks



## Legislative Team Pushes for Federal Support



Savanah Schott meeting with Representative Loeb'sack's office about HR 1849.



Legislative team member Roy Forey (left) at an advocacy breakfast during Rare Disease Week.

It's been a year now since Jim Himes (D-CT) and Ed Royce (R-CA) introduced the HHT Diagnosis and Treatment (DATA) Act HR 1849 in the House of Representatives. This bill would be the first to create a federally led and financed initiative for early diagnosis and appropriate treatment of HHT.

### Here's what the team has been up to (that means you!):

- Secured the support of representatives Zoe Lofgren (D-CA), Rose DeLauro (D-CT), Eleanor Norton (D-DC) and David Loeb'sack (D-IA).
- Reached more than 600 supporters on the HR 1849 PopVox page located at <https://popvox.com/bills/us/114/hr1849>
- Submitted an appropriations request with the National Institutes of Health to ramp up the HHT research plan and sponsor an HHT conference.
- Secured inclusion of vascular malformations to be a part of the Peer Review Medical Research Program for access to additional grants in 2016-2017.
- Attended Rare Disease Week in Washington, DC for advocacy training and events.
- Scheduled more than 20 meetings with congressional representatives to discuss HR 1849 and a sister Senate bill.

**Interested in advocacy?** Join the Cure HHT Legislative Team the weekend of June 17th and 18th for Capitol Hill Day and the DC Walk. Contact [lauren.linhard@curehht.org](mailto:lauren.linhard@curehht.org) or call 410-357-9932 for more info.

**Show your support for the HHT DATA Act** by sending an easy email to your representatives through <https://www.popvox.com/bills/us/114/hr1849>.

## Cure HHT Partners with Anatomy Gifts Registry

*From the bitterness of disease man learns the sweetness of health."*

~Catalan Proverb

Body donation is a generous gift to science and education that allow scientists, clinicians and educators to discover treatments and therapies not possible using animal or artificial models, such as advancements and improvements in the diagnosis and treatment of HHT.

Registration involves a simple pre-screening with consent form indicating one's intent to donate. This allows you to share your wishes in advance with your family. Every case is handled with care by expertly trained coordinators, and if someone does not qualify for the program, alternative options are provided.

Body donation through the AGR program is a no-cost alternative to the traditional funeral or cremation. AGR donors must be:

- 18 or older
- Clear of contagious diseases such as HIV/AIDS, Hepatitis B & C, Syphilis and Tuberculosis
- Reside or pass away within the 48 contiguous United States
- Able to obtain a detailed medical and social history shortly before or at the time of death

Contact AGR directly to request a future donor registration packet or to speak with a caring and compassionate advisor.

### Anatomy Gifts Registry

Anatomy Gifts Registry (AGR) is a nationwide, whole body donation program of the Anatomic Gift Foundation, Inc., a nonprofit organization. Established in 1994, AGR is experienced in procuring, storing and distributing human cells, tissues and organs to researchers and scientists. Tissues are procured through surgery, transplant and autopsy from consented private donors from across the nation.

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**Phone:** (800) 300-5433

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**BECAUSE  
WE KNOW  
ABOUT HHT,  
WE ACT**

# One Simple Action Can Change Lives

Everyday heroes are everyday people, taking a stand to support those who have HHT

## Become a HHT Hero Today!

By creating a campaign on everydayhero, you can help our organization to find a cure  
by hosting an event to raise funds for Cure HHT

What can you do on everydayhero?  
Walk, run, swim, ride, bake, dance, volunteer....  
the options are endless!

Sign up today and find out how YOU can make a difference

Not sure what you want to do, but you'd like to get involved?  
Contact us at [events@curehht.org](mailto:events@curehht.org) and we will help you get started!





# HHT Mom Blogs for HHT Cause

HHT snuck into the Clark family little by little, until suddenly three of the four children and their father were all diagnosed at once with the genetic disease. Wife and mother Desiree Clark turned to HHT stories online to help make sense of the minefield that is living with HHT.

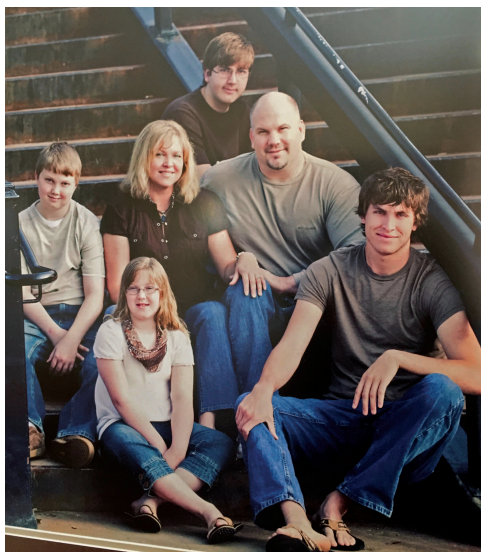


"Watching my family suffer so much was indescribable," Desiree said. "Several times I landed on stories from folks going through the same thing my family was experiencing, so I soaked in every word. It was a comfort to me, in some strange way, that we weren't alone."

Desiree hopes to provide that same comfort as a monthly writer for the new Cure HHT blog, "What the HHT."

Though she doesn't have HHT, she believes her perspective as a caregiver and mother will resonate with many HHT families.

"I'm not a scientist or doctor, so I cannot work on that front, but I can be a face (or fingers for typing) to bring about awareness," Desiree said. "I'm a pretty persuasive speaker and if I believe in a cause I will fight for it. God gave me the talents I have for a purpose, maybe this is it."



**Become a writer for Cure HHT by emailing**  
**[lauren.linhard@curehht.org](mailto:lauren.linhard@curehht.org)**  
**or subscribe to**  
**"What the HHT?"**  
**at <https://curehht.wordpress.com/>**

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## Become a Cure HHT Member today!

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- Access to educational webinar series.
- Reduced registration and VIP reception at Patient and Family Conference
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## The **Cure HHT** webinar series

Knowledge is power, and as a member of **Cure HHT** you are given that power every month with educational webinars designed to empower you as your own advocate. Cure HHT members receive exclusive access to additional educational webinars for only \$55 a year.

Keep an eye on your mailbox and inbox for information about your **Cure HHT** membership opportunity!

**ACT NOW and register for the next Cure HHT webinar at <http://curehht.org/resources/webinars/>.**

“

As an HHT sufferer in Australia, it is easy to feel very isolated. It is nice to know there is support albeit a long way away! Thank you.

- Katherine P.

”

“

The physicians are very thorough and give a good high level description of the challenges faced by HHT patients. The questions posed by the audience shed light on what is most top of mind for the patient dealing with HHT every day.

- Deb D.

”

“

Well worth it and thank you so much. My 28-year-old daughter has just been diagnosed as well, and I hate to see her go through what I have for the last 38 years. This was very informational and gives me hope.

- Sharon T.

”

“

As an HHT patient who has never met another HHT patient, I truly appreciate the webinars and all of the efforts by you at Cure HHT!

- James W.

”

“

I appreciated the review of basic treatments, because I have struggled to find physicians with awareness of the disease, and I also liked that I could participate from the comfort of my home far away.

- Lois D.

”

### **Past Topics Include:**

HHT in Women  
Managing Chronic Nosebleeds  
Cardiac and Liver Manifestations  
AVM on the Brain  
HHT in Children  
Iron Deficiency and Anemia



**Cure HHT**  
PO Box 329  
Monkton, MD 21111



**ACT NOW** by registering for the **Cure HHT Team DC Walk** to bring awareness to our nation's capital. Register and create your fundraising page at <http://2016-dc-hht-walk.everydayhero.do/>.  
**How will you ACT TODAY for Cure HHT?**

If you no longer wish to receive this newsletter please contact [hhtinfo@curehht.org](mailto:hhtinfo@curehht.org) or 410-357-9932.