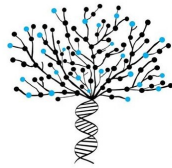


# HCU Herald

**Presented by**



**HCU Network America**

**Connecting for a Cure.**

**There have been a lot of things happening for the HCU community & for HCUNA.  
We strive to keep you informed and connected.**

## HCU Awareness Month has arrived!

**Check out pages 8-10 for ways for fun ways to spread  
awareness for Homocystinuria!**



**GO BLUE FOR HCU**

**October is HCU Awareness Month**

**HCU Network America — [Hcunetworkamerica.org](http://Hcunetworkamerica.org) — [Hcunetworkamerica@gmail.com](mailto:Hcunetworkamerica@gmail.com)**

# Heroes of HCU



Judy



Susie

Judy and Susie Hempling were sisters, four years apart in age, growing up in a large family in Buffalo, NY in the early 1960s. Judy was a very active and pleasant child, but started to have vision issues around age 5, and was also showing signs of a learning disability. Her parents, Ed and Peg Hempling, took her to an ophthalmologist. He told them she needed surgery as she had displaced lenses, but he also recommended they bring her to Children's Hospital in Buffalo, as he had recently become aware of a rare genetic

disease that often first showed symptoms of near sightedness and displaced lenses. (This was in 1964, just one year after Harvey Mudd from the National Institute of Health discovered homocystinuria.).

Judy was tested and diagnosed with homocystinuria (HCU). Her parents, Ed and Peg Hempling, were devastated by the news. They then brought their other children to be tested and received the tragic news that their lovable 2-year-old daughter, Susie, had the disease as well. Judy and Susie didn't seem very sick at the time but they progressively worsened. Pyridoxine (Vitamin B6) was tried but they were not responsive. There was little knowledge back then about protein restricted diets. Their father was especially frustrated because although he was a pharmacist, there were no medications available for their illness.

Both Judy and Susie were considered "mentally retarded" based on their learning disabilities, and attended special education schools. Both suffered over the next several years from blood clots, seizures, displaced lenses, osteoporosis, cognitive deficit, and other issues. Judy had surgery for her displaced lenses and had a stroke during the surgery. They both were hospitalized several times, and while at home often slept in their parents' room as they needed 24 hour care.

Although doctors recommended that moving them to an institution might be the best way to care for Judy and Susie, their parents refused to do so, instead providing them with an abundance of love and the best care and support possible. Even as their disease progressed, Judy and Susie enjoyed life with their parents and five other siblings when they were not acutely ill, including Sunday picnics at the county park, trips to amusement parks, and summer camp at Cradle Beach just outside of Buffalo. They lived for 7 years after their diagnoses, and died within 6 months of each other. Susie died in January 1972 at age 9 from a blood clot in her lungs and Judy died in July 1972 from a stroke. Their story inspired their sister Margie, who was in-between them in age, to do everything she could to help those facing this disease, which led her to form the Hempling Fund for Homocystinuria Research and to work with Danae Bartke to establish HCU Network America.

# HCU and You: Ask Methia

## Dear Methia,

With the holidays approaching, I often stress out about the uncertainty of what is going to be on the dinner table. My parents always make a few items that I can eat if we host the celebration, but when we travel to friends or family members' homes, I can never be sure there will be something I can eat. I also dislike traveling with my formula because having to remember to take it at a social gathering can be very hard. Do you have any suggestions on how to troubleshoot these situations?

Love,  
*Holiday Menu Mayhem*

-----  
Dear *Mayhem*,

It's very understandable to grow anxious around the holidays, with food being central to many gatherings! Traveling and being outside of your comfort zone can make things that much harder. One thing is for certain, though: In order to be successful and compliant throughout the holidays, planning is key. Here are some things you can do to be prepared:

**Take your formula before and after the celebration takes place.** Formula is commonly taken 3-4 times per day. That being said, an event that lasts several hours may span over 1-2 of those dosing periods. You can avoid having to take your formula during the holiday celebration by planning ahead of time. This may mean that your daily formula quantity is split into 2 doses that day instead of 3, or that you set an alarm to remind you to take an extra dose of formula before or after the party begins.

**Ask your geneticist and dietitian about travel letters, or about temporary alternative formulas that "travel well."** Traveling on planes and facing TSA with powdered and ready-to-drink formulas sounds like a nightmare. Many people think that leaving their formula and low protein foods at home is best in order to avoid having to explain or argue with airport security. However, metabolic clinics are happy to write letters to airline security explaining why you must carry your formula and certain foods on the plane with you, since checking it is very risky. Plan ahead of time and ask your clinic to get started on a travel letter for you, indicating the days that you will be traveling and the airline you are flying. If your formula is difficult to travel with, you can also ask your dietitian about a more "compact" formula (in small, individual sachets, perhaps) that might minimize your packing load. Many formula companies are happy to provide samples to patients for these instances.

**On the day of the party, plan to make 2-3 low protein dishes that you can enjoy and share with others.** Scan your favorite blogs, websites, low protein cookbooks, and newsletters (even this one!) for some festive recipes. If you're traveling, ask your host if you can make a quick grocery store run to prepare your favorite vegetable casserole, low protein noodle bake, or compliant dessert. You could even ask them ahead of time if they could have some of these items on hand upon your arrival.

As always, your dietitian is an excellent resource and can point you in the right direction when it comes to formulas, recipes, and suggestions for staying on diet during the holidays!

Sincerely,  
*Methia*

Ask Methia is written by Angela Pipitone, RD, LDN Senior Nutritionist at the McKusick-Nathans Institute of Genetic Medicine within Johns Hopkins University where she works with families affected by metabolic disorders.

# ICIEM and Patient-Expert Meeting in Rio



HCU Network America participated in the 2<sup>nd</sup> International Patient-Expert Meeting for Homocystinuria, which was organized by Tara Morrison of HCU Network Australia as part of the International Congress for Inherited Errors of Metabolism (ICIEM) in Rio de Janeiro, Brazil, in early September.

The Patient-Expert Meeting had over 150 participants, including about 40 patients/family members with HCU from Brazil, along with metabolic clinicians, other health care professionals or researchers involved with HCU. Presentations were delivered by experts in various aspects of HCU from Brazil or globally, on topics including Treatment Guidelines, Quality of Life, Mental Health Issues, Dietary Intervention, Research on New Therapies and support available from Patient Organizations.

## HCU Network America's Role

Margie McGlynn, President of HCU Network America, gave 2 presentations during the meeting. First, she presented an overview of the global research map recently completed for HCU, which outlines research underway to develop new therapies for the disease. These therapies fell into 5 different approaches, which are listed below.

- Enzyme Replacement Therapy to provide a synthetic version to replace the CBS enzyme
- Gene Therapy to deliver DNA to enable the body to produce its own CBS enzyme
- Alternative synthetic enzymes to degrade methionine or homocysteine
- Products to restore faulty CBS enzyme function or activation
- Nutrition or Dietary Supplements to address metabolic imbalances caused by HCU



L to R: Margie McGlynn, Tara Morrison, Kristin Bell



Margie also gave a presentation during the patient-only part of the meeting on HCU Network America and the resources we have available to support patients and family members. The patient stream also included presentations on HCU Network Australia and the Brazilian Patient Association for HCU, as well as a presentation by HCU Network America Board Member Dr. Harvey Levy on the importance of Natural History Studies and Registries for HCU. Also participating in the meeting were Dr. Kim Chapman, Board Member, and James Weisfeld-Adams, Medical Advisor for HCU Network America. A separate physician stream included more in-depth scientific or disease information on HCU.

### Key Take Aways

The key takeaways from the meeting were summarized during the meeting close:

- Early Screening and Diagnosis are critical for HCU

- Aggressive treatment, including dietary management, is critical to prevent clinical symptoms

- Dietary management is key but it can be very difficult to comply with, and better options are needed to improve taste and access for patients

- Mental health disorders need to be recognized, and it is important to control HCY levels to prevent or improve symptoms, and to avoid stigma

- New treatments are key to improve quality of life for HCU patients, and to lessen dietary restrictions. While progress is being made for classical HCU with promising approaches in development (with one expected to start human trials in 2018), similar progress needs to be made for methylation disorders (e.g. MTHFR) and cobalamin disorders.

- There is hope... we are fortunate to have many talented and committed global, national and local experts, many passionate and caring patients and family members, and patient advocacy groups to help bridge the gaps – but we cannot rest until a cure is found for all



# In Case You Missed it!

## Community Call: Fall 2017



# HCU Network America

September 19, 2017 - 7pm Eastern

### Fall 2017 Community Call: Revisited

September 19, HCU Network America had it's Fall Community call. During our call we re-capped work, activities and events that HCU Network America has been active in, including The International Congress of Inborn Errors of Metabolism conference in Rio, engaging companies in helping us improve the landscape of newborn screening, and our clinic contact. During our call we also visited upcoming events such as HCU Awareness Month, our 2018 *Taking the Lead for HCU* conference, among several others.

If you were unable to join us, you can view the recording on our YouTube channel!

### About HCU Infographic

In early September we released our first infographic! Our infographic covers various topics of HCU, including, diet & treatment, affected body systems, how it's diagnosed and associated risk. We hope this will be one of many!

To view the entire infographic, visit our website:

[hcnetworkamerica.org](http://hcnetworkamerica.org) > What is HCU? > Classical HCU > Infographic

### LIVING WITH HCU

facts, figures, and the importance of getting screened at birth

This is Bill.



Hi!

Bill suffers from a rare congenital disease called Homocystinuria, or

# HCU

HCU expresses itself in different ways in different individuals, because of different genetic defects that cause the disease--

--and how well it is controlled via

**diet & treatment**



HEALTHWELL  
FOUNDATION®

When health insurance is not enough.®

### Assistance Reminder

Under this fund, HealthWell can assist with premium costs. Medicare supplemental policies can help with cost shares related to many aspects of your health care. Using a HealthWell grant to cover premiums may be a better option than treatment-specific cost shares. To see if you qualify, please visit:

<https://www.healthwellfoundation.org/fund/homocystinuria/>

# Upcoming Events

## HCU Awareness Month has Arrived!!!



GO BLUE FOR HCU

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### Taking the Lead for HCU

Save the Date ~ April 21 & 22, 2018

**The first HCU Network America Conference**

Westford, Massachusetts

Details to follow this fall at [HCUNetworkAmerica.org](http://HCUNetworkAmerica.org)

Registration opens  
October 15!



## October HCU Awareness Month Activities



Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
Change your Facebook picture to the HCU Awareness Ribbon 1	Start your HCU Facebook fundraiser and invite your friends! 2	Share the HCU Network America HCU Infographic 3	Share a HCU patient story video! 4	Share a pic of your first pair of glasses, or the fact: by age 10, 55%-82% of patients will have their lenses dislocate 5	Share the HCU timeline—if you know other timeline facts, let us know! 6	Share your diagnoses story! 7
#GoBlueforHCU 8	#HCUAwareness post in a public space! 9	Share a low protein meme 10	#Create4Cure create a work of art that brings awareness for HCU 11	Share with one stranger, what HCU is and why it's important to you! 12	Wear jeans for our rare genes 13	Wear your HCU Shirt and share a pic online! 14
Challenge your friends to eat 10 g of protein & a normal protein shake 3 x a day. #ToastToHCU 15	Share a pic of an item that has the same amount of protein you can have 16	Share your completed daily diet record 17	Real cost of HCU: Grocery Cost Comparison 18	Share a pic of what your grocery store "haul" looks like 19	Share your favorite low protein recipe! Bonus if you cook it and share a pic! 20	Dining out low protein style! 21
#HaikuForHCU Write and share a Haiku describing life with HCU 22	Share something you wish people would understand about HCU 23	#High5ForHCU List 5 ways HCU makes you a stronger, better person 24	#Hope4HCU Share 4 things that gives you hope or encouragement 25	#FacesOfHCU Share a pic of you saying "I am one of the 1 in 250,000 people with HCU" 26	Share a pic or video capturing all the medication and formula you take in a day! 27	Share a pic of you and a HCU buddy! Or tag a friend who is a great support! 28
<b>Newborn Screening</b> All 50 states test for HCU at birth, but 50% are missed! 29	<b>Cutting Edge of HCU:</b> Share a HCU Therapy that is in the works! 30	Your message to the world about HCU! 31				

### Notes:

Confused about one of the days activities?

Check out HCU Network America's Facebook page for examples!

Or if you are getting a head start on your planning, email us at [HCUNetworkAmerica@gmail.com](mailto:HCUNetworkAmerica@gmail.com)

**Share your activities on social media and your Facebook fundraising page!**



YouTube



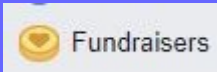
HCU Network America

[www.hcunetworkamerica.org](http://www.hcunetworkamerica.org)  
[hcunetworkamerica@gmail.com](mailto:hcunetworkamerica@gmail.com)

## Day 2 of HCU Awareness Month: How do I create a Facebook Fundraiser?

To create a fundraiser for a charitable organization:

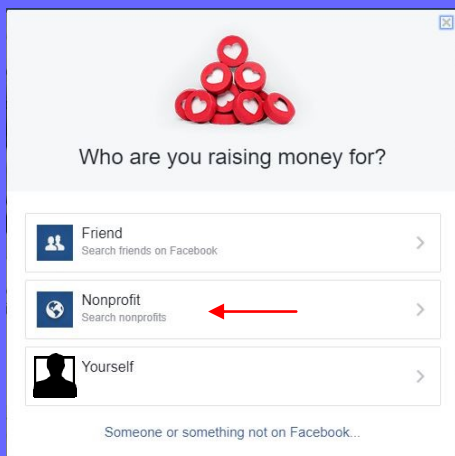
1) Click Fundraisers in the left menu of your News Feed (You might need to select Explore > See more)



2) Click Raise Money



3) Select Nonprofit/Charity.



4) Select a HCU Network America

5) Choose a cover photo and fill in the fundraiser details

We recommend you either make it a picture of the person affected by HCU, or Our HCU Awareness Banner, but in the end the cover photo and details are up to you!

6) Click Create

7) Invite friends to your Facebook fundraiser

8) Share your 31 Days of HCU Awareness social post on your fundraising page!

# Fundraisers

## ❄️ Thirty-One Gifts Fundraiser ❄️ Supporting HCU Network America



**Shop online October 1—October 15**

All orders will directly support HCU Network America.

Wherever you go, go in style. Pack up for holiday travel and save over 60% on one of three totes with every \$35 spent.

Shopping runs from October 1 — October 15

If you want to support HCU Network America and shop for a cause

Email Danae' Bartke for the direct ordering link  
Danaeaustin@gmail.com

## 31 Gifts Fundraiser Benefiting HCU Network America

Start your holiday shopping early and raise funds for Homocystinuria in the process! Shopping starts October 1 and runs through October 15, so start your shopping today!

[Click to Shop now!](#)

## Made to Order Bake Sale

Do you live in the Indianapolis, Indiana area or know someone who does? One of our HCU parents, Amber is hosting a made to order bake sale! She is a professional chef and is taking orders for low protein and high protein baked goods! If you or some one you know is interested in ordering some of her delicious cookies, bars, breads or cheesecakes, please contact her from the information on the flyer!

Amber has already taken in \$100 worth of orders!

Thank you Amber for hosting a fundraiser for HCU Network America!

Made to Order



Items include, but are not limited to:  
Cheesecakes, cookies, brownies, bars, breads

All proceeds will go to HCU Network America  
Samantha was diagnosed with Homocystinuria (HCU) at birth.  
When not treated, Homocystinuria can be life threatening!

To Order contact Amber at:  
Agibson1d@outlook.com or 317-847-5530

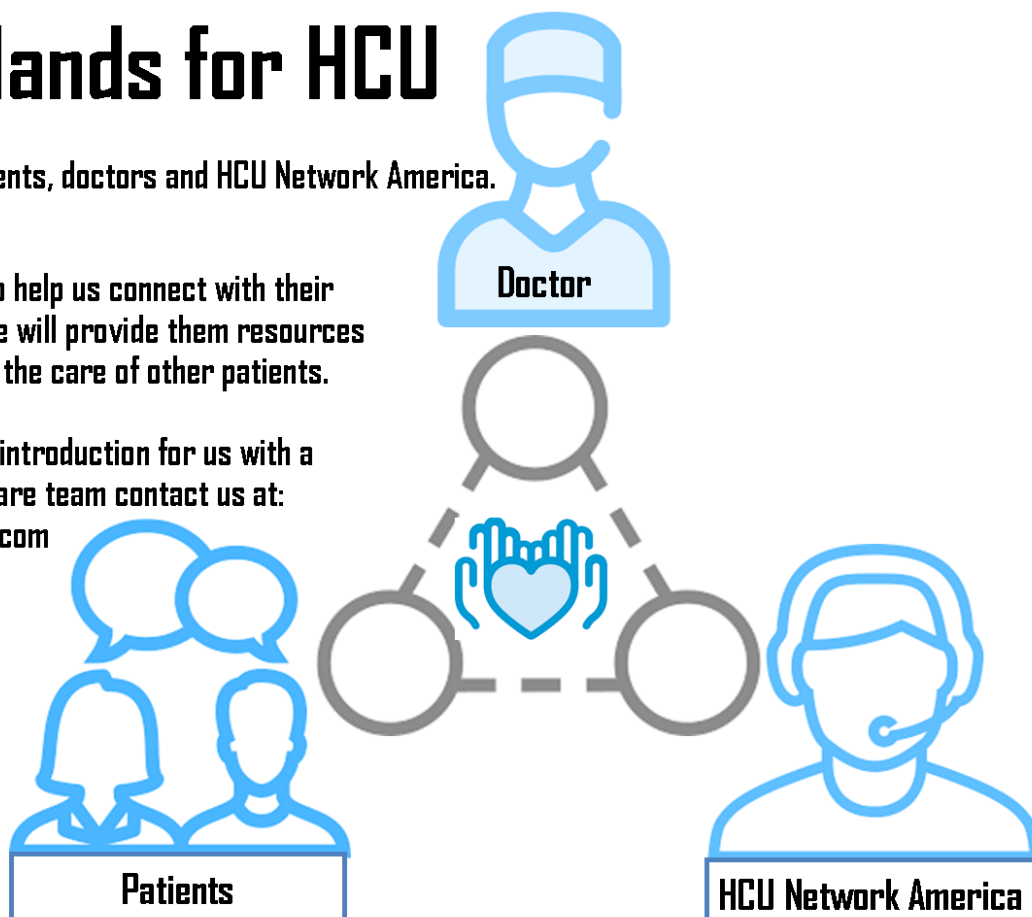
# Ways to Get Involved

## Helping Hands for HCU

A collaborative effort of patients, doctors and HCU Network America.

We are asking for patients to help us connect with their metabolic team. In return, we will provide them resources to help better your care and the care of other patients.

If you are willing to make an introduction for us with a member of your metabolic care team contact us at:  
[HCUNetworkAmerica@gmail.com](mailto:HCUNetworkAmerica@gmail.com)



## Natural History Study

Current sites include: Boston, Philadelphia & Atlanta.

Joining the Natural History Study allows researchers to find out more about Homocystinuria and issues that patients face. Natural history studies help drive new therapies and a cure! If you qualify, we highly suggest you participate if there is a center in your area. You do not have to be a patient at one of these clinics to participate.

[Click here to find out how!](#)

## Amazon Smile

Shop Smile.Amazon.com when purchasing through Amazon and 0.5% will be donated to HCU Network America when you designate us as your charity of choice.

## Company Matching

Did you know that many companies big and small may match donations made to HCU Network America?

**[Click to donate directly](#)**

**We'd like to thank the following content contributors:**

**Editor in Chief: Danae' Bartke**

**Heroes of HCU: Judy and Susie**

**HCU and You: *Ask Methia*, - thanks to Angela Pipitone**

**ICIEM and Patient-Expert Meeting in Rio: Margie McGlynn**

**Fundraiser: Danae' Bartke and Amber Gibson**