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.

Inside this Issue:

Heroes of HCU:
Anniston from Mississippi

Anniston was born in November 2017. We were so excited for our beautiful healthy baby girl. We didn't think anything of the normal routine newborn screening. No news was good news they said as we were leaving the hospital.

About two weeks after she was born we received a phone call from the health department saying one of the tests didn't get enough blood and we needed to bring her in to be tested again. We still didn't think anything of it. Turns out her levels were just at the passing line and wanted to be sure she was okay. When those tests came back the doctors were pretty positive she had HCU. All her pediatrician could tell us was information from an old outdated book. We were referred to a doctor 3 hours from where we lived to confirm. We were heartbroken. All the information we were given at first was so scary. Nobody in our families had ever gone through something like this. It was a lot of information to process. After they confirmed she did have HCU we started seeing a wonderful genetic doctor closer to home. His team has been extremely helpful with helping us understand all this.

After we had come to terms with everything we were struggling to feed Anniston. Pediatrician wrote it off as gas. It eventually got worse and worse. She would refuse her bottles all together. We knew our baby had to eat to develop as a normal baby but she also had this medical problem. She wasn't gaining weight. This took an emotional toll on us. Turns out she had severe silent reflux. Finally 4 months after she was born we got her eating somewhat normal.

We found out early on that Anniston was b6 responsive but every time we would try the b6 it would seem to make the reflux flare up. So her geneticist decided to keep her off it until her reflux was under control. We started the b6 about 4 months ago and so far everything has been going smoothly. We struggle giving her the b6 because it must taste nasty so we have to come up with different ways to give it to her.

Anniston has been allowed 50 mg of methionine a day. She was recently increased to 75 mg. Baby food has been super easy since you know how much methionine is in each tub. Now that she's starting to eat more solids we are learning how to measure all her foods with a scale. We are learning more and more about what foods she can have and how to prepare them. Since she has never been a fan of the bottle it has become harder to feed her the bottle since she discovered foods taste better. We started trying flavor enhancers like strawberry Nesquik and chocolate syrup.

We just celebrated Anniston's first birthday. We are so thankful for newborn screening because our baby girl is able to have a beautiful healthy life. We continue to learn more and more about HCU daily from her amazing genetic doctor and dietitians plus this very supportive community.

HCU and You: *Recipes from the Kitchen*

Strawberry Cheesecake Panna Cotta

Author: Amber Gibson

Servings: 3— 1 panna cotta (about 156 g each)
Protein: 1.9 g
Calories: 337

Ingredients:

- 30 g Cheesecake Pudding, Instant, dry mix only
- 1/3 c (70 g) Sugar, White Granulated
- 2 tsp (6 g) Agar powder
- 13 1/2 fl.oz. (400 g) Full Fat Coconut Milk, canned (Thai Kitchen brand)
- 1 tsp Vanilla Extract
- 32 g Homemade strawberry sauce

Directions

1. Lightly spray three ramekins with cooking spray and set aside.

2. Combine the pudding mix, sugar, and agar powder in a small bowl and lightly whisk to combine. Set aside.

3. Pour coconut milk and vanilla into a small saucepan and place over medium heat. Allow coconut milk to heat up, but not boil. Be sure to stir the coconut milk to avoid burning it. Keep the saucepan over medium heat and slowly add the dry ingredients to the heated coconut milk, using a whisk to stir and avoid any clumping.

4. Continue to cook over medium heat until the coconut milk mixture begins to thicken and coats the back of a spoon, about 2 minutes. Remove from heat and immediately pour into prepared ramekins. Allow to cool to room temperature, then place in the refrigerator to chill for at least four hours.

5. When ready to serve, place strawberry sauce on a small plate. Run a thin knife around the edge of the panna cotta to loosen it from the ramekin. Lightly shake to remove from the ramekin and place on top of the strawberry sauce. Garnish with mint and a strawberry. Enjoy!

Ingredients For Sauce
318 g Strawberries, fresh, slices 60mg
1/3 c Sugar, Brown, packed 3mg
1 tsp Vanilla Extract

Directions:
1. Place all above ingredients in a small sauce pan. Place pan over medium heat and cook while continuing to stir so strawberries won't burn.

2. Cook for about 5-8 minutes, stirring frequently, until the strawberries are cooked and sauce begins to thicken.

3. Remove from heat. Using either a food processor or immersion blender, puree the sauce to desired consistency. Allow to cool. Can be used right away or kept in a glass jar in the refrigerator up to one week.

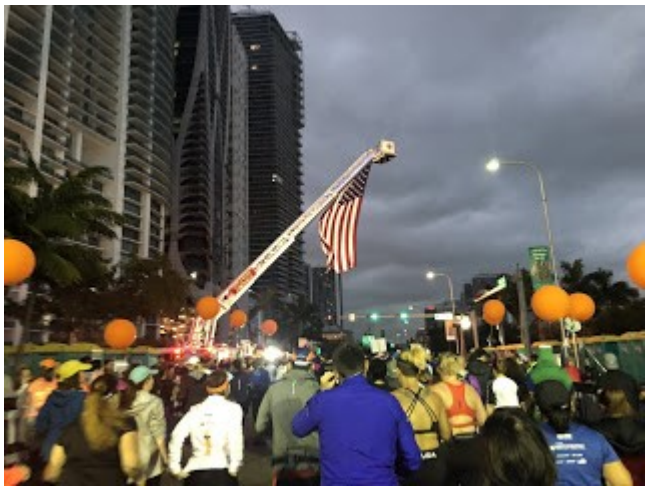
The Rare Runner,
Miami Half Marathon Recap

I am happy to report that the Miami Half Marathon was a success! Here's a quick recap of race day:

I woke up early to get ready, ate breakfast, and head out to the starting line of the race. For breakfast, I had some coffee, a Vitaflo HCU Express20 with water, ¾ of a banana, and a coco banana golden bar. All my race gear was laid out from the night before which made things easy. I attached my race bib to the front of my shirt and a picture of all the HCU Heroes on the back.

The forecast was calling for heavy rain and thunderstorms, not an ideal weather prediction, but at least it wasn't cold. I grabbed a throwaway rain poncho – just in case. My Mom wished me luck before I headed out the door at 5:30am.

The starting line was conveniently just a few blocks from my hotel. On my walk over, I noticed it wasn't raining as predicted and wondered when it would start. I got to the starting line in plenty of time. Although the race officially started at 6am, my corral didn't end up starting until almost 7am. About 15 minutes before my start I consumed a GU energy gel.



Shortly after the race began, I decided to discard my rain poncho. It was a risky move, but it turned out to the be the right one. It was extremely humid, and the poncho was adding an extra layer of warmth that I didn't need. Although, it remained overcast throughout the race, fortunately, the predicted rain never showed up.

The first few miles of the race were past all the cruise ships in the port of Miami. I took in all the sites and snapped a few photos along the way. I slowed down at every water station to make sure I stayed hydrated and consumed additional GU energy gels about every 4 miles.

Along the way, there were more than a few aches and pains, but each time I struggled with the race, I switched my focus to the reason behind my run – the amazing HCU Heroes. Visualizing all those faces and reflecting on their stories really helped me push through the hard parts of the course. When I got close to the finish line, a wave of emotion came over me and I put in one final sprint.

I happily claimed a new shiny medal to add to my collection and met up with my Mom who was spectating the race.

After the race, we headed back to the hotel, so I could quickly refuel with another Vitaflo HCU Express20 and take a shower. After the hotel pitstop, we found an awesome German restaurant nearby that offered some healthy low-pro lunch options. This salad was amazing!

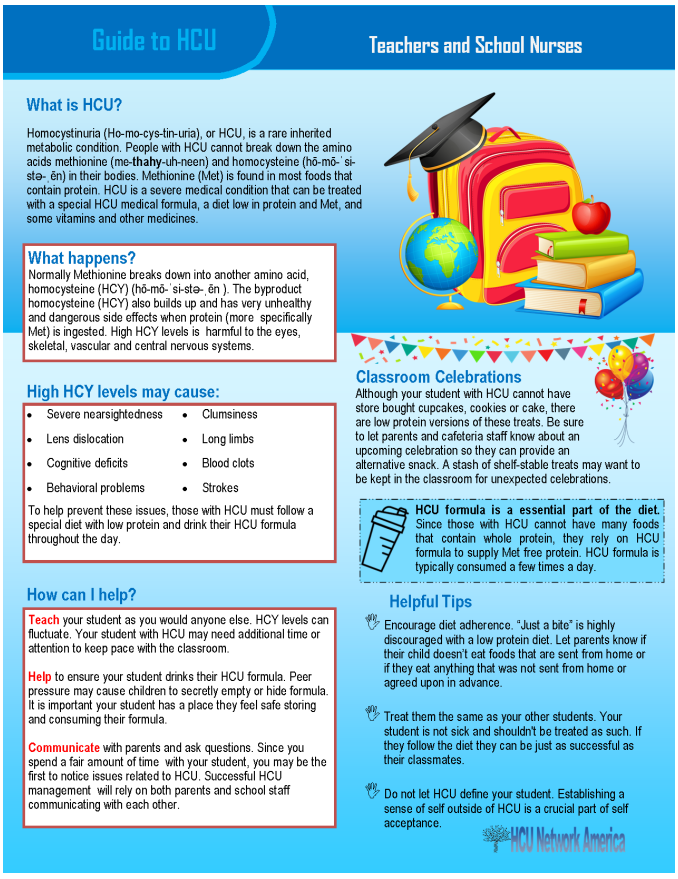
Over lunch, I did what many runners do after finishing a big race - I started planning the next one! After all, it's only January, and my year of running has just begun. Stay tuned for an update on my next race adventure!

Kristin still needs your support!
She may have finished the race, but she still has $3,324 left in her fundraising goal. Help her achieve this goal: **Donate today:** **CLICK HERE**

Educators' Guide to HCU

Is your child in school or going to be starting Kindergarten next year? Ever wonder what information schools should know about HCU ?

Our "hot off the press" Educators' Guide to HCU is designed to spring-board that discussion between you, the school nurse and classroom teacher(s). In our Educators' Guide we give you an easy explanation of HCU, Helpful Tips for Teachers and Nurses, Educational and Nutritional Accommodations tools, as well as ways to ensure students are not left out of classroom celebrations. We know that teachers don't have a lot of time, so this two page guide is the perfect amount of content to help get the conversation started.



Guide to HCU

Teachers and School Nurses

What is HCU?

Homocystinuria (Ho-mo-cys-tin-uria), or HCU, is a rare inherited metabolic condition. People with HCU cannot break down the amino acids methionine (me-**thahy**-uh-neen) and homocysteine (hō-mō- si-stə- ēn) in their bodies. Methionine (Met) is found in most foods that contain protein. HCU is a severe medical condition that can be treated with a special HCU medical formula, a diet low in protein and Met, and some vitamins and other medicines.

What happens?

Normally Methionine breaks down into another amino acid, homocysteine (HCY) (hō-mō- si-stə- ēn). The byproduct homocysteine (HCY) also builds up and has very unhealthy and dangerous side effects when protein (more specifically Met) is ingested. High HCY levels is harmful to the eyes, skeletal, vascular and central nervous systems.

High HCY levels may cause:

- Severe nearsightedness
- Lens dislocation
- Cognitive deficits
- Behavioral problems
- Clumsiness
- Long limbs
- Blood clots
- Strokes

To help prevent these issues, those with HCU must follow a special diet with low protein and drink their HCU formula throughout the day.

How can I help?

Teach your student as you would anyone else. HCY levels can fluctuate. Your student with HCU may need additional time or attention to keep pace with the classroom.

Help to ensure your student drinks their HCU formula. Peer pressure may cause children to secretly empty or hide formula. It is important your student has a place they feel safe storing and consuming their formula.

Communicate with parents and ask questions. Since you spend a fair amount of time with your student, you may be the first to notice issues related to HCU. Successful HCU management will rely on both parents and school staff communicating with each other.

Classroom Celebrations

Although your student with HCU cannot have store bought cupcakes, cookies or cake, there are low protein versions of these treats. Be sure to let parents and cafeteria staff know about an upcoming celebration so they can provide an alternative snack. A stash of shelf-stable treats may want to be kept in the classroom for unexpected celebrations.

HCU formula is a essential part of the diet. Since those with HCU cannot have many foods that contain whole protein, they rely on HCU formula to supply Met free protein. HCU formula is typically consumed a few times a day.

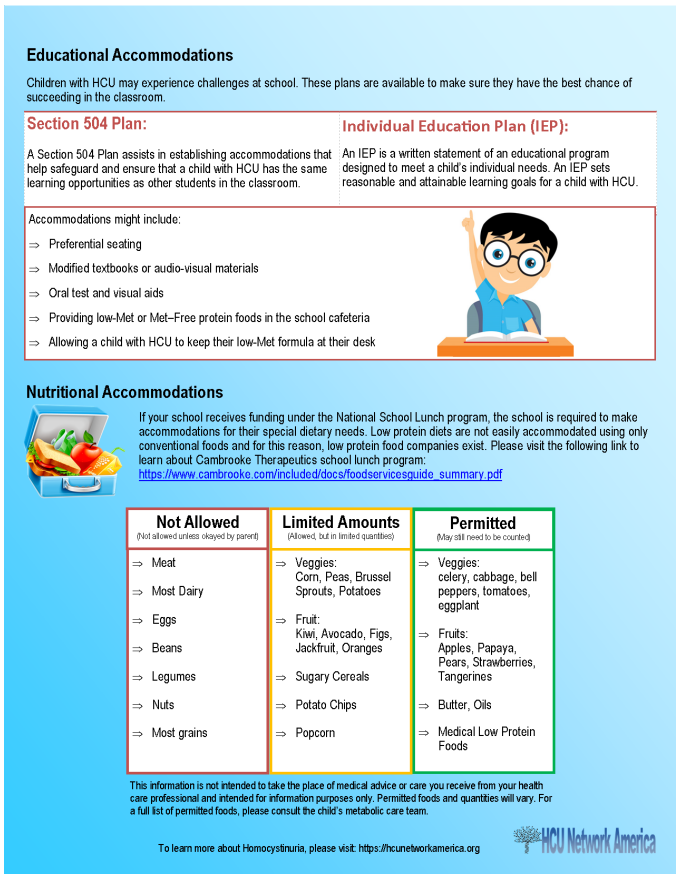
Helpful Tips

Encourage diet adherence. "Just a bite" is highly discouraged with a low protein diet. Let parents know if their child doesn't eat foods that are sent from home or if they eat anything that was not sent from home or agreed upon in advance.

Treat them the same as your other students. Your student is not sick and shouldn't be treated as such. If they follow the diet they can be just as successful as their classmates.

Do not let HCU define your student. Establishing a sense of self outside of HCU is a crucial part of self acceptance.

HCU Network America



Educational Accommodations

Children with HCU may experience challenges at school. These plans are available to make sure they have the best chance of succeeding in the classroom.

Section 504 Plan:

A Section 504 Plan assists in establishing accommodations that help safeguard and ensure that a child with HCU has the same learning opportunities as other students in the classroom.

Individual Education Plan (IEP):

An IEP is a written statement of an educational program designed to meet a child's individual needs. An IEP sets reasonable and attainable learning goals for a child with HCU.

Accommodations might include:

- ⇒ Preferential seating
- ⇒ Modified textbooks or audio-visual materials
- ⇒ Oral test and visual aids
- ⇒ Providing low-Met or Met–Free protein foods in the school cafeteria
- ⇒ Allowing a child with HCU to keep their low-Met formula at their desk

Nutritional Accommodations

If your school receives funding under the National School Lunch program, the school is required to make accommodations for their special dietary needs. Low protein diets are not easily accommodated using only conventional foods and for this reason, low protein food companies exist. Please visit the following link to learn about Cambrooke Therapeutics school lunch program: https://www.cambrooke.com/included/docs/foodservicesguide_summary.pdf

Not Allowed (Not allowed unless okayed by parent)	Limited Amounts (Allowed, but in limited quantities)	Permitted (May still need to be counted)
⇒ Meat	⇒ Veggies: Corn, Peas, Brussel Sprouts, Potatoes	⇒ Veggies: celery, cabbage, bell peppers, tomatoes, eggplant
⇒ Most Dairy	⇒ Fruit: Kiwi, Avocado, Figs, Jackfruit, Oranges	⇒ Fruits: Apples, Papaya, Pears, Strawberries, Tangerines
⇒ Eggs	⇒ Sugary Cereals	⇒ Butter, Oils
⇒ Beans	⇒ Potato Chips	⇒ Medical Low Protein Foods
⇒ Legumes	⇒ Popcorn	
⇒ Nuts		
⇒ Most grains		

This information is not intended to take the place of medical advice or care you receive from your health care professional and intended for information purposes only. Permitted foods and quantities will vary. For a full list of permitted foods, please consult the child's metabolic care team.

To learn more about Homocystinuria, please visit: https://hcunetworkamerica.org

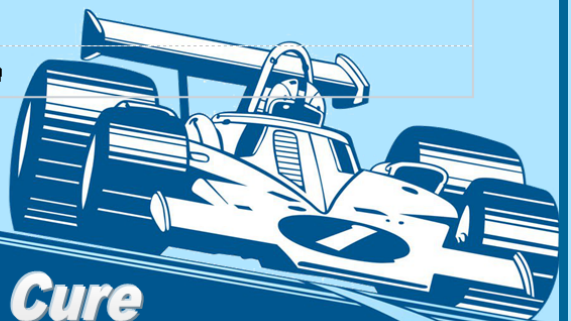
HCU Network America

To download and print the Educators' Guide to HCU
Visit: https://hcunetworkamerica.org/toolkits-and-checklist/

HCU Network America Conference

Agenda At A Glance

Saturday	Topic
7:30—9:00 am	Registration and Vendors Open
7:45—8:45 am	Breakfast
9:00—9:45 am	Introduction and Meeting Expectations
9:45—10:15 am	Vendor Break
10:15—11:15 am	Keynote 1: Natural History Study Update
11:15—12:30 pm	Lunch
12:30—1:00 pm	Vendor Break
1:00—2:00 pm	Breakout Sessions: By age group
2:00—2:30 pm	Vendor Break
2:30—3:30 pm	Keynote 2: Screening: Obstacles, Options and Family Planning
3:30—5:30 pm	Free Time
5:30—8:30 pm	Reception (Dinner Included)
Sunday	**Topic**
7:30—8:30 am	Breakfast, Registration and Vendors Open
8:45—8:55 am	Group Photo
9:00—9:30 am	Vendor Acknowledgement and HCU Hero Award
9:30—10:30 am	Keynote 3: Therapies on the Horizon
10:30—11:00 am	Vendor Break
11:00—12:00 pm	Panel: Ask the Expert
12: 00—1:30 pm	Closing Remarks and Lunch



*Schedule is subject to change.

Accelerating Towards a Cure

2nd Homocystinuria Conference

October 19 & 20, 2019 | Indianapolis, Indiana

Registration is now live. Put the pedal to the metal, register today!

https://hcunetworkamerica.org/2019-conference

Rare Disease Day

About Rare Disease Day

Rare Disease Day® takes place on the last day of February each year. The goal of Rare Disease Day is to raise awareness amongst the general public and decision makers about rare diseases and their impact on patients' lives. It's never too early to get involved!

The National Organization for Rare Disorders (NORD) is the official Sponsor of Rare Disease Day in the United States. To learn what's happening around the globe, visit the Rare Disease Day website at rarediseaseday.org.

What is a Rare Disease?

In the U.S., any disease affecting fewer than 200,000 people is considered rare. This definition comes from the Orphan Drug Act of 1983 and is slightly different from the definition used in Europe. There are more than 7,000 rare diseases affecting 25-30 million Americans. In other words, one in ten Americans are suffering from a rare disease and more than half of them are children.

Besides dealing with their specific medical problems, people with rare diseases struggle to receive a proper diagnosis, find information and get treatment. The rarity of their conditions makes medical research more difficult.

Show Your Stripes™

The zebra is the official symbol of rare diseases in the United States and is noted for its black and white stripes, which are central to its uniqueness. Everyone has his/her own stripes, those characteristics that make each individual distinct. While each of the more than 7,000 rare diseases are unique, there are many commonalities that unite patients, families, caregivers and supporters. In the spirit of raising the profile of the rare disease community at large and celebrating Rare Disease Day, this year NORD is promoting a variety of ways in which individuals, organizations and groups can "show their stripes."

Raise Social Awareness

HCU Network America will be sharing a fact each day about Homocystinuria on our social media accounts. We encourage you to share our post.

In addition to sharing our post, we encourage you to check out the NORD Social Media Toolkit

Rare Disease Day - Casual for a Cause

Itching to forgo the traditional slacks and skirts for a more relaxed look at the office? Take the lead in your organization or company to sponsor a Casual Cause: Jeans Day to raise funding for the resources and tools HCU Network America provides to the Homocystinuria community! Encourage employees to dress down for a day or even an entire week by requesting donations in exchange for a day in their casual best.

Not sure where to start? Not a problem! We've created several "Team Captain" materials to help you on your way toward sponsoring a successful fundraiser. Take a look at our customizable flyers, stock emails to inspire participation, tracking sheets and more! View resources at: https://hcunetworkamerica.org/casual-for-a-cause/

For more information, please contact Danae' Bartke at 630-360-2087 or dbartke@hcunetworkamerica.org

We'd like to thank Christie Norenberg at The First Interstate Bank, Kalispell branch in Montana for helping organize Casual for a Cause Day Jeans day for HCU Awareness Month. Because of her efforts, they were able to raise $240!

What does Rare meant to you?

During February, leading up to Rare Disease Day, we challenge you to write an Acrostic Poem about Rare means to you.

An acrostic poem is a poem where certain letters in each line spell out a word or phrase. Typically, the first letters of each line are used to spell the message, but they can appear anywhere.

Remember to use the hashtags #ShowYourStripes , #RareDiseaseDay #GoBlueforHCU

Here is our example:

What does rare mean to you?
Rarely recognized by its name
Always needs more Awareness
Requires low protein diet
Explanations guaranteed

#Homocystinuria
#ShowYourStripes , #RareDiseaseDay #GoBlueforHCU

Rare Disease Day 2019: HCU Gear!

Get your Rare Disease Day gear before time runs out! Last day for guaranteed devliery by February 28th is February 8th! https://bonefire.com/rare-disease-day-hcu

VMP, LLC
expanding genetic horizons...

Voice: 404.793.7800
Fax: 866.744.5665
www.vmpgenetics.com

HELP US TEACH PHYSICIANS ABOUT HOMOCYSTINURIA

FACT! Teaching about metabolic diseases in medical school and residency programs is poor.
FACT! Most patients live and die without a diagnosis being made, especially when the disease presents in adulthood.
FACT! Patients cannot access effective therapies unless a proper diagnosis is made.
FACT! The sooner a diagnosis is made and treatment begun, the better the outcome.

WE NEED YOUR HELP!

We at VMP Genetics believe in the power of "patient-teaching" and are bringing patients and families into lectures and presentations - at conferences and in the classroom around the country. While doctors teach facts, patients tell stories. Story-telling is a more compelling teaching method with better recall over time than didactic lecturing. We also believe that doctors are more likely to make a diagnosis if they have already seen a patient and heard her/his story. Story-telling can be live or taped...

WE ARE LOOKING FOR...

- ***Patients and/or family members who are interested in telling their stories in local medical classroom settings...*** We are developing a Patient Teacher Registry. If a medical school faculty member is looking to introduce the patient story in a teaching session, the Registry can tell him/her if there are patient-speakers in the area and what diagnoses they have.

- ***Patients and/or family members who are interested in having their stories videotaped...*** As we secure funding, we are interested in recording stories that reflect the broader patient experience. The more variety in the stories, the richer the learning potential.

- ***Videos of patients and families telling their stories...*** A 5-or 10-minute clip can be downloaded into a lecture about that disease or relevant biochemistry to enhance the learning potential of the session.

Please help us in our efforts to raise awareness about Homocystinuria through this innovative educational outreach to the medical community. To Volunteer or participate, or for more information about this project... please contact Jacob Athoe at PatientTeacherRegistry@gmail.com

Mark Korson, MD
VMP Genetics
Director of Education

Jacob Athoe
Genetic Counseling Student
Boston University Genetic Counseling Program

OT-58, Enzyme Replacement Therapy Clinical Trial Recruitment

Orphan Technologies has initiated a first in human (Phase 1) clinical trial of OT-58, an enzyme replacement therapy that addresses the underlying enzyme deficit for patients living with classical homocystinuria. The goal of this trial is to evaluate the safety and efficacy of OT-58 in patients with classical homocystinuria and identify the appropriate dose. Patients between the ages of 12 and 65 years of age with classical homocystinuria may be eligible to join. For additional information on criteria for eligibility, please go to:

https://clinicaltrials.gov/ct2/show/NCT03406611?cond=Homocystinuria&rank=1

There are four sites in the US currently participating in the trial:

- Children's Hospital of Philadelphia – open to patient enrollment
- Boston Children's Hospital – open to patient enrollment
- Indiana University – open to patient enrollment
- Children's Hospital Colorado - open to patient enrollment

Payment for time and travel may be available to patients who participate in this trial.

To inquire about participation into the trial, please email: info@orphantechnologies.com

Contact Register

What is the contact register?

The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission to. By registering, you will be also be able to identify other affected patients in your state and request their contact information, and you will be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?

HCU Network America strives to inform and provide resources for patients and families, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission - plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://hcunetworkamerica.org/contact-register/

We'd like to thank the following content contributors:

Editor in Chief: Danae' Bartke
Heroes of HCU: Anniston from Mississippi
HCU and You: *Recipes from the Kitchen: Amber Gibson*
Educators Guide to HCU: Patient—Parent Advisory Committee
Casual for a Cause: Website content: Fundraising Committee

Click to donate directly