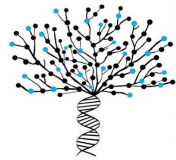


HCU Herald

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.**

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Contact Register



Heroes of HCU:

Elliott from South Carolina



July 10, 2018 was the worst day of my family's life. This was the day that our sweet Elliott, 2 years old, was put into a medically induced coma and placed on the ICU Floor of the children's hospital where he would remain for 29 days. Elliott was having seizures with no outward signs and doctors could not figure out why. They would later find out that the seizures were a result of a series of blood clots in the veins throughout the brain, which also resulted in Elliott having a stroke. I will never forget the words spoken to us by the doctor on July 15, 2018 – "We want you to know how serious this is. We don't expect to lose Elliott to this, but you need to know that we could." It would be what seemed like an eternity (but was really just 11 days after Elliott was admitted to the ICU) that doctors were able to pinpoint a cause for everything. "Homocystinuria", they told us. It took a doctor looking at Elliott's 'whole picture' and realizing that his homocysteine levels were through the roof (250). This diagnosis was a blessing for us because it meant having an answer. But, this diagnosis was the beginning of a new lifestyle and a huge learning curve for our family.

Elliott is now home and doing well! He is on lots of medications and a brand new diet. We are counting the methionine in his foods and he is allowed to have 126 mg each day. In addition, he must drink his HCU/Prophree shake daily.

How has the journey with HCU affected our family so far? Well, mostly it is hard to be spontaneous. No hopping in the car and going out to eat just yet. Our lives involve a great deal of planning now. We have to make sure that anywhere we go, we have Elliott's medications and either food that has already been weighed or we must bring our scale. We are also navigating through the uncertainty of feeding a semi-picky toddler who is very limited by what he can eat. Since we are counting methionine and that is not something that is readily posted on food and drink packaging, we are 'playing it safe' right now by only giving him foods where that information is available. Our new lives also (at least for right now) involve a lot of worries and unknowns. We wonder why Elliott's homocysteine level jumps from week to week. We worry when he doesn't feel good or isn't acting himself – could it be just a normal pain or a common cold? Or is he acting funny because there is something wrong internally that we cannot see? It can be very scary, especially since this is all so fresh and new to us.

If I could give some advice to a family just starting their journey with Homocystinuria, I would say to first, take a deep breath. This new diagnosis will come with a lot of changes. It will be a lot to take in at first. But, you can do it! And knowledge is power. Now that you know what you're dealing with, you can help yourself, your child or your family member to live a healthy life! It will be a learning curve, but there are plenty of people and resources to help you along the way! For us, it was the Facebook group "Homocystinuria awareness and support" where we have been able to connect with other families. Also, the HCU Network America has some great resources and even held their very first conference this year! Most importantly, you are not alone.

HCU and You: Recipes from the Kitchen

Amber's Veggie Loaf

By Amber Gibson

Makes about 7 loaves

Each loaf is about 1.12g protein

Ingredients:

1 20 oz can jackfruit in brine, rinsed well and squeezed dry
114g button mushrooms, about 5
40g diced onion
1 clove garlic, minced
40g finely shredded carrots
1/4 cup Cambrooke Burger Mix
1 Tbsp Coconut Aminos
1 Tbsp Ketchup
1/4 cup nondairy creamer (I use Coffee-Mate)
salt and pepper to taste



Directions:

Preheat oven to 350 degrees. Spray a mini loaf pan with cooking spray.

Put the jackfruit and mushrooms in a food processor and pulse until crumbly. Move mixture to a medium bowl.

Add the rest of the ingredients to the jackfruit mixture. Use a spatula to combine.

Using a cookie scoop, put two scoops of the loaf mixture into each mini loaf cavity and spread evenly to fill each cavity.

Bake at 350 for 30 minutes. Allow to sit in pan for 5 minutes before serving.

Back To School With Amazon



**Shop at AmazonSmile
and Amazon will make a donation to HCU Network America!**

Do you shop on Amazon? If so, you can have a portion of your eligible purchase price come to HCU Network America! Instead of logging into the normal Amazon website, simply type this link into your browser for the portal to the AmazonSmile for HCU Network America.

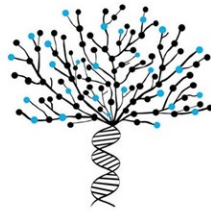
<https://smile.amazon.com/ch/81-3646006>

Amazon will donate 0.5% of the price of your eligible AmazonSmile purchase to **HCU Network America** whenever you shop AmazonSmile. AmazonSmile is the same Amazon you know. Same products, same prices, same Amazon Prime benefits – but it helps us!

After your initial visit to the link above, you can go directly to [Smile.Amazon.com](https://smile.amazon.com) (make sure you bookmark this site) and your continued shopping will benefit us.

Please visit and encourage others, too!

Rare Bears



HCU Network America

Rare Science - Accelerating Cures for Rare Kids, is a non-profit focused on raising awareness and funds to support development of therapies for children with rare diseases, partnered with HCU Network America to deliver Rare Bears to children with HCU in celebration of National HCU Awareness Month in October! A Rare Bear is a grassroots community-driven, hand-sewn one-of-a-kind teddy bears for one-of-a-kind “rare” kids made by volunteers working with Rare Science.

To order your bear, follow the link: <https://www.rarescience.org/hcu/>

Deadline to order is September 21st!

Natural History Study

Orphan Technologies is pleased to announce that it is expanding its Natural History Study Sites!

Orphan Technologies has just signed an agreement with Children’s National Medical Center (Institution) in Washington, District of Columbia and Children’s Hospital Colorado in Aurora, Colorado to include them in the Natural History Study.

The study does not involve any investigational medications, but will provide information to researchers who are currently developing a medication to treat the disease.

The study has 5 main parts, all of which are provided free of charge to participants, bone exam (DXA scan), blood draws for important health tests, comprehensive eye exam, cognitive and physical exams.

Other sites include:

Emory University, School of Medicine Department of Human Genetics
Decatur, Georgia

Indiana University School of Medicine
Indianapolis, Indiana

Boston Children’s University
Boston, Massachusetts

The Children’s Hospital of Philadelphia
Philadelphia, Pennsylvania

To learn more about the study, please visit the link below:

<https://clinicaltrials.gov/ct2/show/NCT02998710>

New Board Member: Mark Lewis

Meet our Newest Board Member, Mark Lewis



Mark and his wife Karen's first-born child Benjamin was diagnosed with Homocystinuria through the Massachusetts New Born Screening Program in July of 1991. This diagnosis came as a complete shock to them and their pediatrician because of the rarity of the disorder. They immediately brought Ben to Boston to a Metabolic Specialist. Needless to say, it was devastating news as parents, but after they took the time to learn about HCU they knew that it was up to them to provide Ben with the best chance of living a healthy, happy life through a special low protein diet and medical formula. Mark and Karen went on to have two more children, both girls, Chloe and Gabbi. Chloe was tested at birth and did not have HCU. Gabrielle on the other hand was the first baby diagnosed through amniocentesis. Hearing a diagnosis of HCU the second time around was not any easier or less devastating than the first time, but now they were better prepared. Ben and Gabbi have excelled both academically and professionally. All three of Mark's children have

grown to be kind and compassionate people, partly as a result of living with HCU. Aside from the personal aspect of having two children diagnosed with homocystinuria, Mark pursued the need to become involved with the metabolic community. Mark's involvement happened very early on after Ben's diagnosis. As time went on, his focus shifted primarily to attending to his family's needs while working in the medical device and drug development industries. Mark is currently the Vice President, Program and Outsource Management at TARIS Biomedical. Mark agreed to join the board of HCU Network America after being a very involved participant at our first patient/family conference in April, and he is hoping to apply his professional knowledge and personal experience to help support HCUNA in achieving our goals for the betterment of those challenged by HCU and related disorders.

Fundraising & Patient-Parent Advisory Committee Formation

At our inaugural conference in April, we closed the conference by asking you, the community, for input on moving forward. We asked the community what their most important needs were, what we should focus on going forward, as well as what volunteer opportunities you'd be interested in. From your feedback, we went back home and put our heads together, and we were able to form two committees that will benefit the HCU community greatly: a Fundraising Committee and a Patient-Parent Advisory Committee.

We created a mission and goals for each committee, which are a work in progress and will evolve as our community grows. We then formed committees based the list of those who volunteered and chose committee chairs who we felt would be strong leaders for each committee. You will find descriptions of each committee and their background information on the following pages. Thank you all for agreeing to serve!

Fundraising Committee

Fundraising Committee

Mission: To create effective strategies and tools to help HCUNA increase success in fund raising to support our operating costs and help engage and support the HCU community to create and carry out fundraisers. Over time, help develop strategies and programs to support fund raising to support research grants.

Goals/Responsibilities:

- 1) Create templates for fundraisers that the community can do
- 2) Engage volunteers, help get them started with their fundraisers
- 3) Be a point of contact and resource for those with fundraisers or new ideas
- 4) Develop ideas for HCU Awareness Month that can be done locally or on-line and help HCUNA communicate and engage the community
- 5) Longer term, help develop strategies and tools to raise funding for research grants



Committee Co-Chair: Cole Sullivan

Cole is the father of two kids with Homocystinuria. His daughter Colbie was diagnosed after she suffered a blood clot in her brain and some small strokes shortly after her 3rd birthday. After he and his wife learned that Homocystinuria is a genetic disorder, their son Cayle tested positive and was officially diagnosed two months later. Since the diagnosis of both children, Cole and his wife have spent their time networking with other patient families, studying the diet that goes along with HCU, and trying to learn as much about HCU as they can. Cole holds a degree in Business Management and resides in the state of Montana with his wife, Sarah, and their two children.

Laurie Bonucci is also serving as a committee co chair—please see her bio on next page.

Meet the Patient– Parent Advisory Committee

Mission:

The Patient-Parent Advisory Committee will come together to provide advice on how best to support the needs of the HCU community.

Goals/Responsibilities:

- 1) Provide ideas to HCUNA on what would be helpful to the HCU community, e.g. through periodic calls with the executive director and other board members as needed
- 2) Act as an ambassador to new HCU families - providing them with information, tools, and advice
- 3) Review draft content and materials for new resources being developed by HCUNA and provide feedback
- 4) Help serve as or organize volunteers as needed for events such as the next Patient-Expert meeting (registration, t-shirts, goody bags, etc)

Patient-Parent Advisory Committee Co-Chairs: Laurie Bonucci, Karen Lewis and Sarah Sullivan.



Laurie is married to Steve and they farm in Northern Illinois. They have 4 adult children. Joe, their second son has HCU and Type 1 Diabetes. Laurie worked on the Chicago Mercantile Exchange as a commodities trader before children and after having kids; she volunteered in her community. Her works includes Princeton Library Board of Trustee for 13 years, Illinois Valley Community College Trustee for 4 years, and Master Gardener among many others. She has a Bachelor of Art degree from Lake Forest College and a grant writing certificate from Northwestern University. Laurie is getting her 200 RYT certification in yoga teaching in October 2018. She hopes to bring her energy and enthusiasm to HCU Network America and serve in any way she can.



Karen Lewis and her husband Mark were first introduced to Homocystinuria when their first-born child, Benjamin, was diagnosed as a result of the Newborn Screening Program in Massachusetts. Needless say it was devastating news as parents, but after they took the time to learn about the disorder they decided to reach out to families with PKU for support since there weren't any other families they knew with HCU. It quickly became clear that there was a great need for a parent support group and Karen and a few other PKU parents formed a non-profit group called the New England Connection for PKU and Allied Disorders. It was comprised of parents and professionals, Dr. Harvey Levy at Boston Children's (and a board member for HCUNA) being one of the founding members. Karen held several positions in the organization and remained on the Board for many years. She has two daughters as well, Chloe and Gabbi. Her youngest, Gabbi was also born with HCU and as life got busy with the three children, she focused less on

NECPKU to put all her attention on the kids and Mark. Karen has been a stay at home mom since Ben was diagnosed and feels that now that the kids are getting older, it would be a good time to get back into doing what she can to help other new HCU parents and families. She understands the positive impact it can have on a family to be able to talk to a mom who has been through the difficulties and joys of raising a child with HCU. It brings her such pride to see parents and patients reach out to both Ben and Gabbi to ask them questions about growing up with HCU, and see how well they are doing socially, academically and professionally. Karen's greatest joy in life is being a mom to her three children. Being able to help other families and have a positive impact in the HCU community is a wonderful bonus.



Sarah is the mother of two kids with Homocystinuria. She is a Registered Nurse in the state of Montana. Her daughter Colbie was diagnosed after she suffered a blood clot in her brain and some small strokes shortly after her 3rd birthday. After she and her husband learned that Homocystinuria is a genetic disorder, their son Cayle tested positive and was officially diagnosed two months later. She hopes that by sharing their story and being an active member in the HCU community it will bring more awareness and make newborn screening better.

We are still looking for people to join our fundraising and patient-parent committee's.
If you are interested, please contact us at hcunetworkamerica@gmail.org

Newborn Screening Awareness Month

Newborn screening is a very special topic to our community. We are grateful that all 50 states test for Homocystinuria at birth, but that doesn't mean we don't have anything to worry about. It is estimated that 25-50% of patients are still missed by our newborn screening methods. Because so many patients are still missed we are working diligently to change the current system.

How can you help?

Newborn screening survey

Regardless if you were diagnosed at birth or later in life, please have your geneticist fill out our newborn screening survey and submit them to Dr. Ficicioglu or on our website. You can find the survey at:

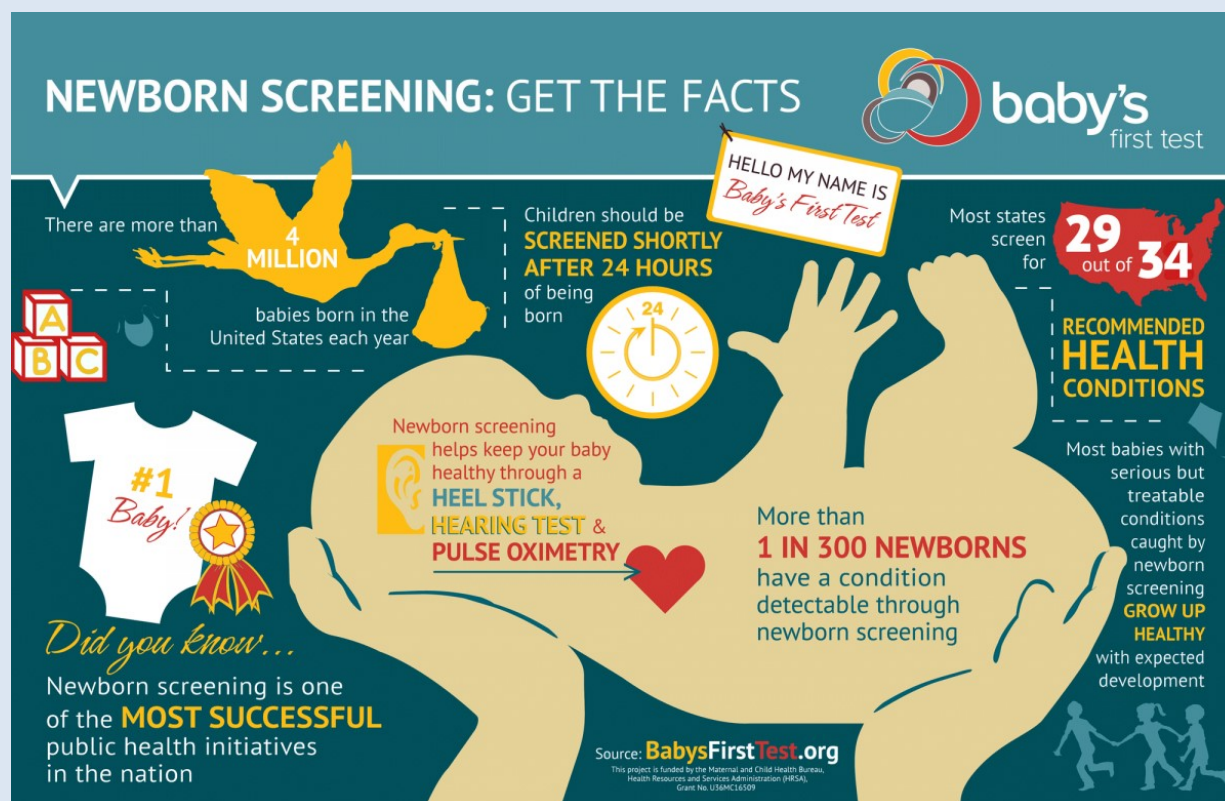
<https://hcunetworkamerica.org/survey-on-classical-homocystinuria-patients-missed-by-newborn-screening/>

Share your story

Share your patient story how newborn screening has impacted your life, or how it would have, had you had proper newborn screening. Use the hashtags #myNBSstory #NBS18 #homocystinuria

Share your story with HCU Network America— we will then feature you on our website and social media pages!

Email us your story at: hcunetworkamerica@gmail.com



HCU Awareness & You

GO BLUE FOR HCU

October is HCU Awareness Month

HCU Network America — Hcunetworkamerica.org — Hcunetworkamerica@gmail.com

Thanks to an anonymous donor, any funds you help raise from October through December 31 will be matched up to \$20,000!

Start planning your Homocystinuria Fundraiser now!
Have a fundraiser planned? Email us and we will help spread the word.



Need an idea? <https://hcunetworkamerica.org/fundraising/>

Design a shirt, or other item and sell it online—we will share the link!

Backyard Carnival (Perfect for a Halloween Event!)

Walk/Run, 5K or Marathons

Restaurant Night (Chipotle, Mod Pizza, McDonalds are some examples)

Need help? Email us - we will guide you through the process.

HCU Awareness Month Calendar

HCU Awareness Month Activities

Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
	1) Change Facebook picture to the HCU Awareness Ribbon	2) Start your HCU Facebook fundraiser & invite your friends	3) Share the HCU Network America Infographic	4) Share a HCU patient story video!	5) Share a pic of your first pair of glasses, or the fact: by age 10, 55% -82% of patients will have their lenses dislocate	6) Share the HCU timeline - if you know other timeline facts, let us know!
7) Share your diagnosis story!	8) #GoBlueForHCU	9) #HCUAwareness post in a public place!	10) Share a low protein meme	11) #Create4Cure create a work of art that brings awareness for HCU	12) Share with one stranger what HCU is & why it's important to you!	13) Wear jeans for our rare genes
14) Wear your HCU Shirt and share a pic online!	15) Challenge your friends to eat 10 g of protein and a normal protein shake 3 times a day. #ToastToHCU	16) Share a pic of an item that has the same amount of protein you can have	17) Share your daily diet record - completed	18) Real cost of HCU: Grocery Cost Comparison #MedicalNutritionEquityAct	19) Share a pic of what your grocery store "haul" looks like	20) Share your favorite low protein recipe! Bonus if you cook it and share a pic.
21) Dining out, low - protein style!	22) #HaikuForHCU Write and share a Haiku describing life with HCU	23) Share something you wish people would understand about HCU	24) #High5forHCU List 5 ways HCU makes you a stronger, better person	25) #Hope4HCU Share 4 things that give you hope and encouragement	26) #FacesofHCU Share a picture of you saying "I am one of the estimated 1 in 250,000 people with HCU"	27) Share a picture or video capturing all the medication and formula you take in a day!
28) Share a picture of you and a HCU buddy! Or tag a friend who is a great support!	29) Newborn Screening: All 50 states for HCU at birth, but many patients are still missed	30) Cutting Edge of HCU: Share about a HCU therapy that is in the works!	31) Your message to the world about HCU!			

Confused about one of the activities?

Find examples and resources on our website: <https://hcunetworkamerica.org/hcu-awareness-month/>

Follow us on Facebook, Twitter and Instagram during the month for examples and additional activities!

Help spread awareness, share your participation on social media and your Facebook Fundraising page!

Navigating our Future

Global Research Map Update

October 23, 2018—7pm EST

Featuring, Margie McGlynn, HCU Network America President

Over the past few years, a project was sponsored by HCU Network America and HCU Network Australia to develop a global research map that describes the research underway for new therapies to treat homocystinuria, focusing on HCU caused by CBS deficiency. A global research strategy was also developed and Scientific Advisory Board was convened to define the top priorities for funding via new global grants program. This presentation will summarize the findings from this project and status of the global grants process and the status of trials underway.

For more details and to register: <https://hcunetworkamerica.org/global-map-webinar/>



2019 Rome Conference

rome
2019 28 FEB - 1 MAR

3rd International Patient - Expert Meeting



See the link for more info: <https://www.hcunetworkaustralia.org.au/patient-expert-meeting-2019/>

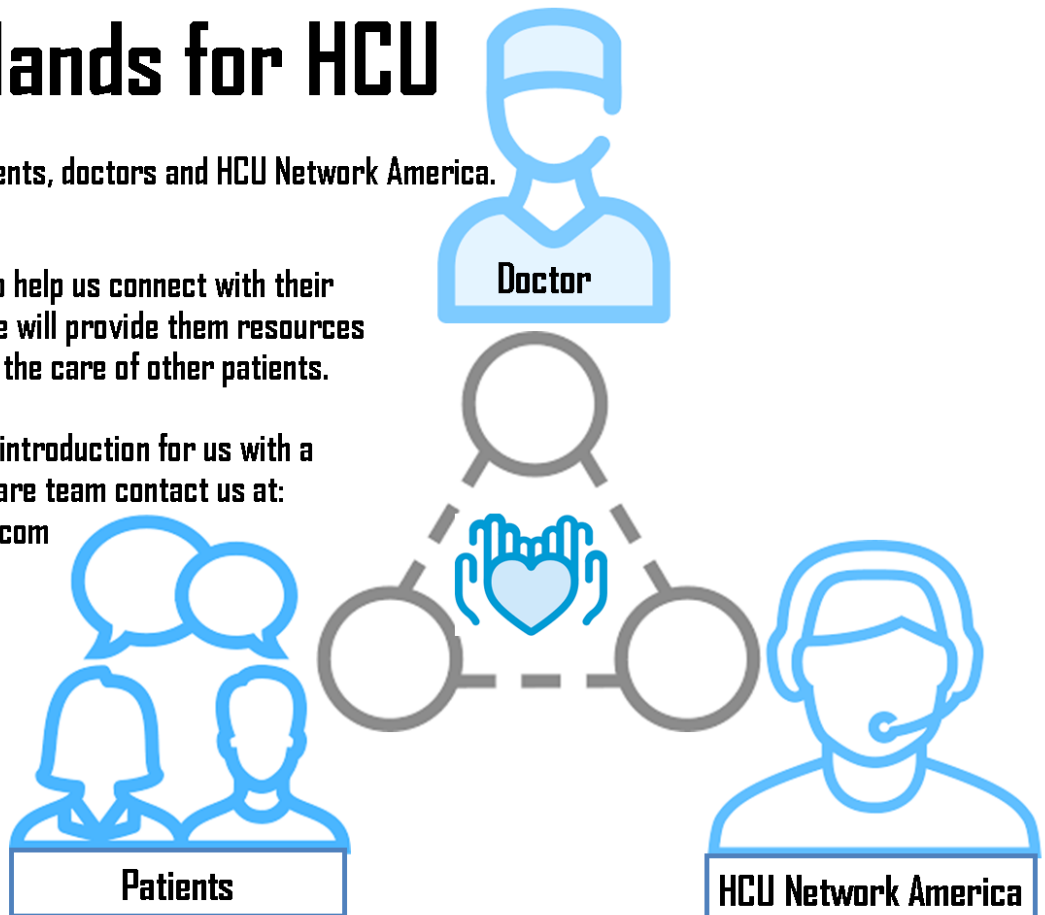
Ways to Get Involved

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



HCU Network Australia Patient & Caregiver Survey

experience of
diagnosis
and
patient
satisfaction.



PATIENT & CAREGIVER SURVEY

There is currently limited evidence showing the considerable length of time taken in some cases to reach a diagnosis of homocystinuria and the level of patient satisfaction regarding access to information, treatment options and medical care. This survey is important to highlight the current situation and has been prepared in consultation with a Key Opinion Leader (KOL) in Europe together with input from HCU Network America, to ensure the information gathered can be used globally to help support improved care for all HCU patients.

We ask no matter what your experience, good or bad, you complete the survey.

To complete the survey click [here](#).

Contact Register



Did you know that just under one year ago we launched our 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/>

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.

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

Company Matching

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

[Click here to find out more](#)

[Click to donate directly](#)

We'd like to thank the following content contributors:

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