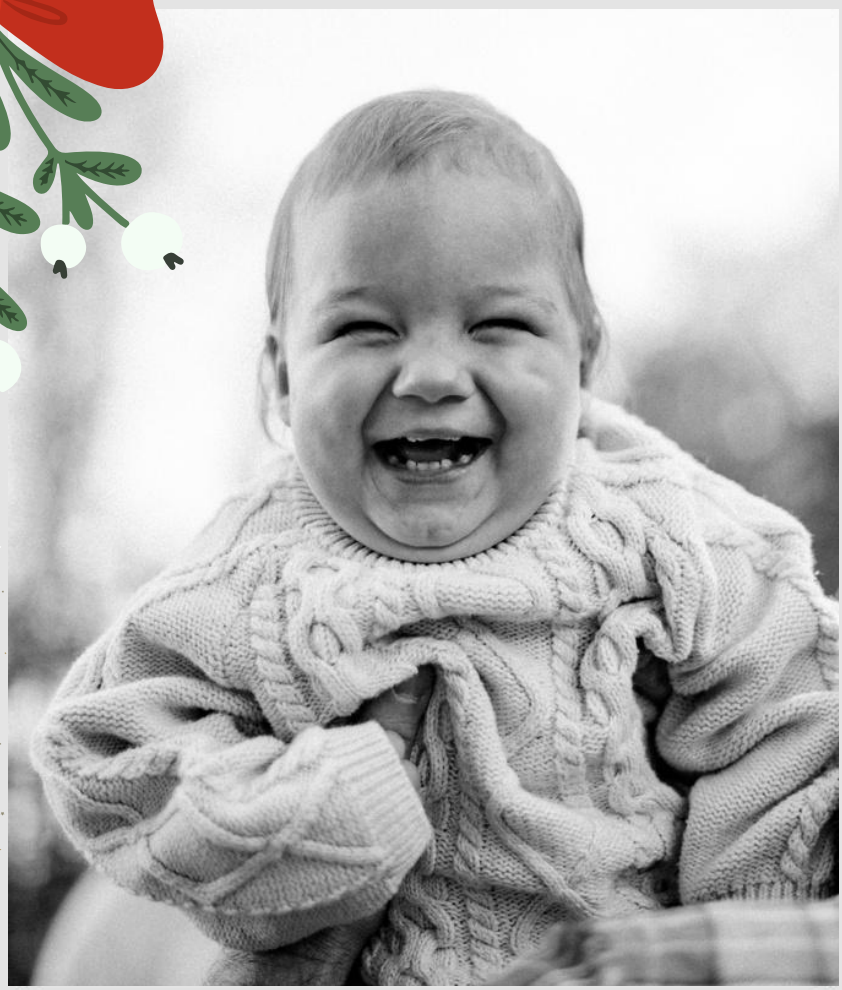
The HCU Herald

Featuring...



HCU Hero
Elijah from Ohio

December 2024

All things Homocystinuria: patient stories, resources, research, events and more!

HCU HERO: ELIJAH FROM OHIO

Elijah was born June 2, 2023. We opted not to do genetic testing because no one in our immediate or extended families had any known genetic diseases, and our first child had been born healthy. Elijah did not get diagnosed until he was about 13 weeks old.

We first noticed when Elijah was five weeks old that he wasn't gaining as much weight as he should have been and was a lot sleepier than is normal. We were admitted to our local Children's Hospital soon after that for failure to thrive. We were in the hospital for five days and Elijah was discharged with an NG tube. Over the next month we worked tirelessly to help him gain weight, but his weight gain wasn't at the typical rate and his condition continued to get worse. He continued to have feeding issues, became more lethargic, and was throwing up four to eight times per day. He was also having diarrhea, occasional blood in his stool, and started developing purplish splotches on his skin. When he was awake, he was very irritable.

Despite bringing up these concerns with our PCP, they did not want to check any blood work and thought what he was experiencing was because of his poor feeding. As a mom and a nurse, it's hard to feel like you are trying to advocate for your child, but also want to respect the judgement of a doctor who has been in practice for over 30 years and is well regarded professionally. At the time, we were also in the process of changing to a new pediatrician's office because the one we had been going to got a new job out of state and would be leaving the area soon. Our GI team initially thought Elijah may have a milk protein allergy, so I cut dairy out of my diet. There was also some confusion about who we needed to see about his care and ended

HCU HERO: ELIJAH FROM OHIO

up essentially being told to follow up with our GI team at Children's because it was a "feeding issue". There was a lot of back and forth and countless prayers as I felt that something wasn't right and didn't feel like anyone was truly listening to my concerns.

When we saw our GI doctor at Children's, on August 24th, 2023, she brought up his skin color, which seemed 'off' to her, and asked me what I thought of it. I was so thankful for her validating my concerns and ordering blood work. The first specimen that was collected clotted and they called me a few hours later to tell me to get labs redrawn whenever it was convenient for me. My gut told me something was wrong, so we immediately went to our local children's lab and had them re-drawn. They called me around 9 pm to tell me that Elijah's hemoglobin was critically low, at 3.9, and to take him to the emergency department right away. With it being so late in the day, Danny stayed home with our daughter, Norah, and I took Elijah in.

As a nurse who has administered blood transfusions to children for a hemoglobin of around 6, this was terrifying, but I also figured that it must have been dropping slowly. When we were discharged from the hospital the first time, his hemoglobin was at a low-normal range. Based on my unofficial calculations, when I first brought up his skin color to his PCP, it is likely that his hemoglobin was around 6, which would've been a transfusable level. Initially I felt stupid for not having taken him when I felt like I wasn't being heard, but it is also different when it is your kid and you're trying to trust the judgement of people with more knowledge and expertise than you. It was a good reminder for me to always trust my gut.

HCU HERO: ELIJAH FROM OHIO

The nurses in the emergency department tried everything possible and could not get an IV going. They tried nine or ten times (with heat packs, ultrasound and the vascular access team etc) and eventually the attending doctor had to do a femoral stick just to get labs. We were sent to the ICU sometime after midnight as a "just in case" until the Children's central line team was in later that morning. Fortunately, nothing eventful happened and a PICC line was placed without any complications. Because Elijah's anemia was unusual, the Hematology team recommended genetics labs prior to receiving blood transfusions. Prior to the blood transfusions, his hemoglobin went down to 3.1.

Elijah got three blood transfusions, and they literally brought him back to life. He went from being sleepy and irritable to being alert and content. As his lab work started to trickle in, we found out that he likely had an "inborn error of metabolism", and it was pointing in the direction of a very rare B vitamin metabolism problem.

As you can imagine, we were shocked. We met with the genetics team while we were in-patient and scheduled a follow up to discuss our genetic testing results. Thankfully, Elijah was doing well enough with the treatments we had started that we were able to recover at home while waiting for definitive results.
Genetic testing took a couple weeks to come back, and it was finally confirmed that he has Cobalamin E Deficiency. We now know that Danny and I are both carriers and that the carrier rate is estimated to be 1 in 300,000. Since this is an autosomal recessive disease, we have a 25% chance of having a child with this disease. Elijah is one of an estimated less than 40 people in the world with CblE.

Since diagnosis, Elijah has progressed tremendously. He has always been behind on milestones but is catching up. We've gotten to really appreciate and enjoy each stage a little longer. In a lot of ways, he is not too far off target when we consider month zero as the month when we started full treatments. He goes to a combined

HCU HERO: ELIJAH FROM OHIO

PT/OT/speech therapy session for two hours each week and early intervention comes to our house 1-2 times per month. He is now 16 months old and is crawling everywhere and pulling to stand. We can tell he is making more neural connections with how his playing and babbling has changed and improved. He had said "mama" a few times, but it was never clear if he was saying it about me. Yesterday a friend was over, and she was holding Elijah and asked him if he wanted her to hold him or if he wanted mama. He replied "mama" and reached towards me. My heart about burst. Brief snapshots of the last year flew through my head and I was overcome with gratitude for how far he's come.

We also recently participated in HCU Network America's Race for Research and Norah, Elijah's older sister, named our team "Team Butterstick" after a nickname she gave Elijah (which is hilarious to me given that he's always been in about the 1st percentile on the growth chart). Danny ran with him in the stroller and did the rest of a Murph workout with Elijah in an Osprey Pack on his back. We had fun and raised way more money than we ever thought we would. We are looking forward to next year and already brainstorming how we can get our community more involved. More importantly, we are looking forward to what can be researched and improved in terms of treatment and quality of life for kids with rare homocystinuria/cobalamin diseases.

What is Cobalamin E (CblE) deficiency?
CblE is a rare inherited metabolic disorder in which the body is unable to properly process vitamin B12 (cobalamin) due to a deficiency in a protein called TCBL1 (transcobalamin B), which is responsible for transporting vitamin B12 into cells. If not diagnosed and treated early, Cobalamin E deficiency leads to neurological problems, anemia, and can cause developmental delays.

How is CblE diagnosed?
Diagnosis involves blood tests showing elevated levels of homocysteine. Genetic testing may confirm mutations in the gene responsible for transcobalamin B.

How is CblE treated?
Hydroxocobalamin, Betaine, Folinic Acid

HCU HERO: ELIJAH FROM OHIO

I thank God regularly that Elijah was born when he was, that other families have gone before us and graciously shared their knowledge and experience, and that modern medicine gives him a chance at life. Elijah is such a sweet and happy baby, and it has been so incredible to see his personality come out. It hasn't been an easy journey at all, but I am thankful for how it has brought our family closer. It's helped Danny and I to be a better team, and our families have been incredibly supportive. I am also grateful that I've been able to connect with other amazing families through HCU Network America. Our stories and experiences are similar and different in their own way, but we still understand each other on a level that few others can fully comprehend. I look forward to seeing how our kids forge their own path and become the amazing people who they were meant to be.

Click here to read Elijah's & other patient stories on our website!

Raising Joy Double Good

FUNDRAISER

RESULTS

HCU NETWORK AMERICA

Raising Joy Double Good

Total Sales	$1237.00
Total HCUNA Donation	$618.00
Stores With Sales	5
States Reached	9
Supporters	24

Thank you to all who participated by hosting a sale or purchasing popcorn!

These funds will be applied towards our End of Year $25K match!

HC&U Podcast

Meet your hosts!

Welcome to the HC&U Podcast!

We are Ben and Lindsey, your hosts. We are so excited to host this podcast as extra resource for the Homocystinuria community. We hope you like our content!

You can read Ben's patient story here.

The latest episode

Ben welcomes Holly, who recently shared her husband Chris's story as our November HCU Hero feature! Ben & Holly will discuss Chris's long journey to a diagnosis, and the role that she played in achieving an accurate diagnosis after many years of setbacks. We'll also get an update on how Chris is doing now!

Missd Chris & Holly's story in our November newsletter? Read it here!

To Listen:

https://hcunetworkamerica.org/hcu-podcast/
or click below on your favorite option!

LISTEN ON Spotify | LISTEN ON iHeartRADIO | Listen on Apple Podcasts | Listen on amazon music

MEET CHRIS AND HOLLY

a story of hope in the face of misdiagnosis

“Serious, but treatable”

is a phrase often used to describe conditions like homocystinuria (HCU). When identified early, HCU can be managed with treatment, allowing individuals to live typical lives. Unfortunately, approximately 50% of patients are missed by newborn screening, resulting in delayed diagnoses and sometimes life-threatening consequences. Two of the most common symptoms of untreated homocystinuria are blood clots and strokes. Individuals with these symptoms are often first seen by ophthalmologists and hematologists, where the correct diagnosis of homocystinuria may be missed.

Today, we want to share a story that embodies resilience, determination, hope, and the profound impact of a misdiagnosis — Chris and Holly’s story.

When Holly met Chris eight years ago, she quickly learned about his complex medical history. Chris had been diagnosed by a hematologist with a blood clotting disorder and elevated homocysteine levels.

Chris’s journey began at the age of 14 with his first deep vein thrombosis (DVT). By his early 20s, he had suffered a massive stroke due to delayed medical care and misdiagnosis. This stroke left him with significant brain damage, vision issues, and a host of other health problems.

HCU NETWORK AMERICA

Over the years, Chris faced multiple DVTs, venous stasis ulcers (non-healing open sores on the lower leg), and severe anxiety, all while being misdiagnosed with bipolar disorder.

Holly, with her clinical background, knew something was amiss. She spent years researching and advocating for Chris, confronting specialists and demanding better care. Her persistence paid off when she finally got Chris the correct diagnosis: classical homocystinuria. This rare metabolic disorder had been the root cause of his elevated homocysteine levels and numerous health issues.

With an accurate diagnosis, Holly's unwavering advocacy resulted in the introduction of new medications and dietary changes that reduced Chris's homocysteine levels from a life-threatening 450 to a manageable 39. He no longer needed numerous psychiatric medications and began feeling like a completely new person, as if a heavy fog had lifted. This transformation has given both Chris and Holly hope for a healthier future.

Chris's story is a testament to the importance of accurate diagnosis and the power of advocacy. It highlights the critical need for continued research, education, and support for those with homocystinuria.

HCU Network America acknowledges the significant gap in education within the field of hematology which can

lead to misdiagnoses for patients with homocystinuria. To address this, we have developed a Hematologist Toolkit designed to assist hematologists in identifying the symptoms of undiagnosed homocystinuria and facilitate referrals to genetic specialists.

We are also excited to announce that HCU Network America will be participating in the American Society of Hematology's (ASH) 66th Annual Meeting and Exposition in San Diego, CA. Our primary objective at this event is to bring awareness to hematologists and educate physicians on recognizing the signs and symptoms of homocystinuria, as well as to offer guidance on patient referrals to genetic experts. We are looking forward to building relationships with the over 30,000 attendees from over 100 countries in attendance! Additionally, we are in the process of creating a guidebook specifically for patients diagnosed in adulthood or later in life, to help them navigate the complexities of the medical system, and to educate them on the treatment for HCU.

As we approach the end of the year, we kindly request your support to continue our mission. Your generous donations enable us to provide essential resources, support, and hope to families like Chris and Holly's. Together, we can make a meaningful difference in the lives of those affected by homocystinuria.

“Chris's story is a testament to the importance of accurate diagnosis and the power of advocacy. It highlights the critical need for continued research, education, and support for those with homocystinuria.”

Matching Gift Challenge

Three generous donors have pledged to match EVERY gift up to $25,000 of donations!

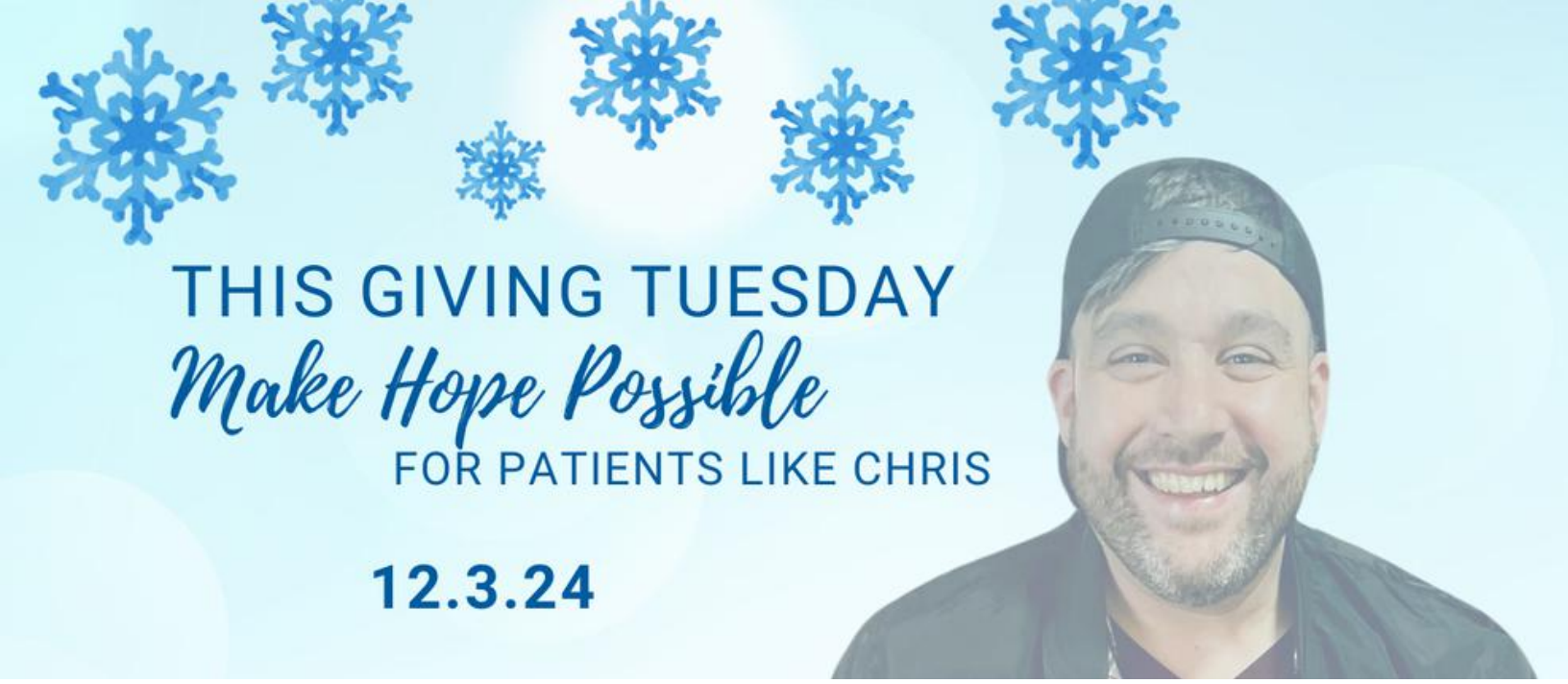
During the winter holiday, warm hearts and generosity can be felt near and far. We ask every patient and family to help us raise funds to continue our homocystinuria community outreach, education, and advocacy initiatives to educate providers and be there for families like Holly and Chris'. Please share our appeal letter with your colleagues, friends, and family.

Three Easy Ways to Donate

Text text the code

"HCU2024" to 44-321

By check
HCU Network America
15 S. Mallory Ave
Batavia, IL 60510



What is Giving Tuesday?

It's a global generosity movement unleashing the power of radical generosity. GivingTuesday was created in 2012 as a simple idea: a day that encourages people to do good. Since then, it has grown into a year-round global movement that inspires hundreds of millions of people to give, collaborate, and celebrate generosity. *This year, Giving Tuesday will be* ***December 3.***

How do I get involved?

We're asking you to assist us in reaching our $10,000 GivingTuesday fundraising goal.

To get started:

- Set up your own GivingTuesday fundraiser on GoFundMe, Facebook, or Instagram!
- Start to reach out to your friends and family in advance and get them to pledge a donation on December 3rd!
- Let them know that by giving to HCU Network America, they are supporting programs and resources that directly benefit the patients and caregivers of our community.
- Remind them that *all donations* are tax-deductible AND will be matched by 3 anonymous donors (up to $25,000!)

Keep an eye on HCU Network Amrica's social media on 12/3!

We will be posting *"12 reasons to give this Giving Tuesday"*, which will feature all 12 of our 2024 #HCUHeroes!

The posts will include a link to our donations page, so if you'd prefer, you can share our posts instead of setting up your own fundraiser.

GIVING TUESDAY

EMPLOYER MATCHING GIFT PROGRAM

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?
Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?
Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?
If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately $45,000 in corporate matching gifts!

How Do I Find out if my Employer has a Corporate Matching Gifts Program?
Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

Top Matching Gift Companies

Company	Match Ratio
General Electric	1:1
Gap Corporation	1:1
ExxonMobil	3:1
Johnson & Johnson	2:1
Microsoft	1:1
Pfizer	1:1
Coca-Cola	2:1
And many more!	

Did you know some companies match retired employees donations?

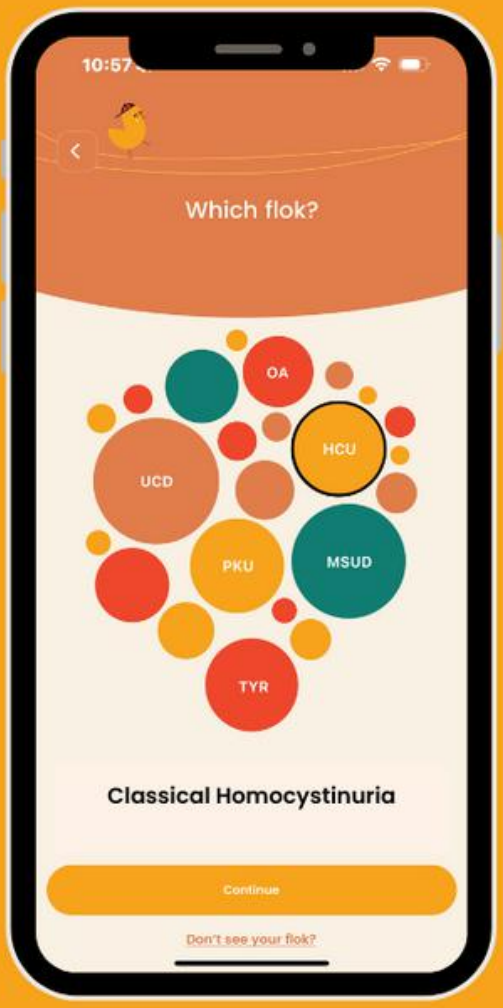
Start using the flok app today!

Welcome to the next generation of metabolic care for the CLASSICAL HCU COMMUNITY

The flok app is now in Open Beta in the United States.

Download at flok.org/app

flok



Big news!

The flok app is now in Open Beta, available for download to everyone in the U.S.!

Jump in and start exploring - the app helps you manage your diet and build a full picture of your metabolic health - including symptoms & moods, activity, medications, and lab tests.

UPCOMING EVENTS

iGive.com

You Shop. Your Charity Gets Money. For Free.

Your holiday shopping makes a difference

Shop at any of the 1,700 stores that want to help, and **HCU Network America** will receive a portion of your purchase, ranging from .5% to over 20%! Stores pay for it all. Never pay more, and sometimes less with coupons and deals.

Want to make your purchases count?

The optional **iGive Button** is a simple web browser app, easy to install and uninstall. It automatically activates at participating stores.

Don't want the Button or an app? Just start your shopping trips by going to iGive.com.

Shop normally (no special codes, no special anything) at any of about 1,700 stores. The Button is working in the background to let them know you're helping when you shop.

Ready to shop?

Head to igive.com to sign up! Make your first purchase within 30 days and iGive will donate a bonus $5 to HCU Network America! Happy shopping!

Walmart · HSN · QVC · KOHL'S · L.L.Bean · macy's · 1-800 flowers · Booking.com · Clarks · GAP · Shipt · & more!

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RARE X

Heart and Blood Vessels Survey, and Blood/Bleeding Survey

In the 1985 publication "The Natural History of Homocystinuria due to Cystathionine β-synthase Deficiency" by Dr. S. Harvey Mudd et al., a study with 629 patients he and his colleagues reported by age 15 years without treatment that 12 % of B6-responsive and 27% of non-responsive will have thromboembolic events.

Does this still hold true? You tell us!

Complete the Survey
https://rare-x.org/homocystinuria/

CANPKU+ CANPCU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

Now Enrolling

Acappella

Sponsor: Travere Therapeutics
Study type: Natural History (no investigational medicine given)
Study duration: About 6.5 years
Goal: To learn more about classical HCU & the course of the disease

TO QUALIFY*	AGE OF PARTICIPANTS	DETAILS
Diagnosis of Homocystinuria due to CBS Deficiency (Classical Homocystinuria)	Currently enrolling 1 to 4 years old	The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

**Your child will need to meet all other study criteria to take part in the ACAPPELLA Study.*

Study Locations
United States: Colorado, Washington DC, Georgia, Pennsylvania
Countries outside of the US: Ireland and Qatar

Approximately 150 people aged between 1 and 65 will participate at sites in the US, Europe and other countries around the world.

150

The ACAPPELLA Study has already enrolled 100 adults and children over 5 years old, and is now looking for children aged 1 to 4 years old to take part.

You may be able to receive payment for time and travel if your child participates in this study.

Talk to your doctor and family members about your child joining the ACAPPELLA study.

Sites are open and currently enrolling participants. For the most current information about the ACAPPELLA Study and to see additional eligibility criteria, please visit:

https://www.clinicaltrials.gov/study/NCT02998710

If you have any questions, please email:

medinfo@travere.com

For more information, please scan the QR code or visit:

www.hcuconnection.com.

MA-PE-24-0008. Version 2.0; en-US; March 2024

TRAVERE THERAPEUTICS

Sign our NBS Screening Petition!

In this petition, we call for state newborn screening labs to **revise screening protocols for Classical Homocystinuria** to *ensure fewer false negative screening results and delayed diagnoses.*

Click here to sign the petition!

Get your kit!

Our FREE Customizable Kits are here!

Request yours today!

At HCU Network America, we believe that one of the most important steps to empowering patients and caregivers is giving them the support and tools needed to succeed! We know that a new diagnosis can be overwhelming and riddled with concerns and questions. To us, one way to combat those feelings, and give you the confidence you need, is by providing you with one-on-one support, educational resources, and practical tools, such as scales, cooler bags, and more! Our request for a kit survey allows you the opportunity to request a one-on-one introductory call (with more opportunities to connect), and then a customized kit to the patient's needs. Don't want a call or a Zoom? That's fine too - we are happy to send you the customized kit.

Request your kit now - **https://www.surveymonkey.com/r/HCUKitSurvey**

**Kits can only be sent to patients in the continental US. However, we are happy to connect virtually and share the educational materials with you via weblinks*!

Stay connected: Join our Contact Register!

What is a contact register?

- a secured private survey that allows you to share information on you or your family member with HCU with us. (general contact info, diagnosis, etc)
- kept confidential and will not be shared unless you permit us

Why join?

- subscribe to our monthly newsletter and other communications
- identify other patients in your state and request their contact information
- access information posted over time that can only be shared with the patient community
- helps us plan events and inform the development of resources and educational tools
- supports our advocacy efforts and enables us to have informed conversations with doctors, pharmaceutical companies, state newborn screening labs, and lawmakers.

I want to participate! What's next?

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://www.surveymonkey.com/r/HCUContact

HOPE CONNECTS US

FOLLOW US