The HCU Herald

September is Newborn Screening Awareness Month!

Featuring...

HCU Hero
Carter from Georgia

September 2024

HCU NETWORK AMERICA

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

HCU HERO: CARTER FROM GEORGIA

Carter was born in February of 2024. He was ten days late, but he was a perfect little baby; everything that we'd hoped and prayed for. His first 24 hours of life were pure bliss; almost like a dream. I'd had a very easy pregnancy and birthing experience, which to me almost felt too good to be true. Carter did have a little bit of jaundice, but other than that, everything was just fine. After a few routine days in the hospital, we went home and life as a family of three began.

Things were pretty quiet for the first couple of weeks until we received a phone call from the pediatrician's office. The person on the line, I believe it was one of the nurses, told us that that something in Carter's newborn screening results came back abnormal and we needed to come into the office. When we sat down with Carter's pediatrician, he said that the newborn screening test showed an elevated level that could indicate a rare genetic disorder called homocystinuria. But, he gave me hope that this was not the case. He told me that in the 13 years that he's been a doctor, he has never had an infant or child diagnosed with this disorder, and that it's not uncommon to send a baby back for follow-up testing. But, deep down in my heart, I somehow knew in my heart that Carter was going to have this disorder. Throughout my entire pregnancy, I had this unexplainable feeling that something was going to go wrong like I was just waiting for the other shoe to drop. So, even as the doctor tried to reassure me, I just knew.

The next thing he told us was that they couldn't perform the blood test in office, so we would need to head to the hospital's lab, which is exactly what we did.

HCU HERO: CARTER FROM GEORGIA

Within a day or so, we got a phone call back from the pediatrician. Carter's homocysteine level was 29; not horribly elevated, but enough that the pediatrician wanted to refer us to a geneticist. While we waited, I did what every parent in my shoes would do: I turned to the Internet. I need to figure out what we might be up against - what is homocystinuria and what would this mean for our future? I searched Google, YouTube, Instagram, and Facebook for anything I could find on HCU. The things that scared me the most were blood clots, strokes, brain damage, eye damage, possible death if left untreated, and the low-protein diet! It was a lot to take in.

Once we connected with the geneticist, he had Carter go in for another blood draw to check the status of his homocysteine level. We went in for the blood draw and got a phone call the next day. Carter's homocysteine levels had doubled from 29 to 60. My husband and I had genetic testing done; a saliva swab test along with blood work, and that is what confirmed our fears: Carter did indeed have classical homocystinuria. As the geneticist explained the disorder to us, it sounded a lot like what I had found in my research. I knew that the fact that Carter had been diagnosed early likely meant that we could avoid complications from the disorder, so my worries in that moment went straight to the idea of the low-protein diet that Carter would have to follow for the rest of his life. My mind was spinning. I needed to hear from real people. That's when I found HCU Network America, and I began to read patient stories which gave me some relief. To know that my son wasn't alone and that there were a group of people who are going through the same things we are, and that there is hope for a normal life for my boy was reassuring.

"I knew that the fact that Carter had been diagnosed early likely meant that we could avoid complications from the disorder, so my worries in that moment went straight to the idea of the low-protein diet that Carter would have to follow for the rest of his life."

HCU HERO: CARTER FROM GEORGIA

When Carter was six weeks old his geneticist started him on liquid B6, which only brought his numbers down a few points, so we doubled it. I was concerned; nothing scared me more than high homocysteine. His levels got as high as 98, and that's when I got really scared (Dr. Google said that a homocysteine level over 100 was dangerous.). The B6 did bring his levels down (to around 70), but not as much as we would have liked. So, it was determined that Carter was partially B6 responsive, and we'd need to try some other things to get his levels down further. We started Betaine when he was 3 months old as well as Hominex-1 formula. Since then, Carter's levels have gone all the way down into the 20's! We are so thankful for that.

Our sweet boy is now 6 months old and thriving - he's meeting all milestones, and making our hearts melt each and every day! He does really well taking the formula. When I first smelled it, I thought "there is no way he's going to eat this!", so I was pleasantly surprised that he has taken to it so well. We're about to start introducing solid foods, so we're already looking at protein content and trying to learn which foods are better options than others. I'm not going to lie: the low-protein diet worries me. As Carter gets older, how am I going to keep him from foods that he can't eat? How am I going to make sure he isn't given food by a family member who forgets or doesn't know what he can and can't have? What about when he goes to school and kids try to switch lunches? (we all know how kids are!) What the heck am I going to feed him? It seems like he can't have anything! What do I say to him when he questions why his parents eat all these foods he can't? My heart breaks that this is his reality for the rest of his life.

HCU HERO: CARTER FROM GEORGIA

As difficult as navigating this new reality seems, I can't express how happy I am that there is a community for families and patients with HCU. With how rare this disorder is, I assumed that there wouldn't be many resources out there, but I've already discovered so much. There's a lot of support out there, you just have to look for it. For example, I found a mom on Instagram who shares the low-protein lunch ideas that she makes for her child. I thought "ok, that's doable! I can do that." In addition to connecting with families with HCU, I've followed some PKU families to get their perspectives and ideas on low-protein foods. I also saw that HCU Network America just held a family conference, and we can't wait to attend conferences in the future to meet families like ours!

Looking back at our journey so far, I feel a sense of gratitude for the timeliness of Carter's diagnosis. As much as a wish he did not have a rare disorder, I am so thankful that it was caught at newborn screening (I've since heard that it's believed that 50% of babies with HCU are missed by newborn screening!). I remember getting a brochure in the hospital with information about newborn screening and letting us know that Carter had undergone the screening. I took it home and put it aside. It wasn't until we got that phone call saying that Carter's screen was abnormal that I got the brochure out and really read it. I didn't understand at the time how important that screen was, but I sure do now.

My hope for is that Carter can lead a normal life and not feel like he's different from other people just because of the way he has to eat and the formula he has to take. I also hope that the newborn screening for HCU can be improved so that other babies are able to get a diagnosis and start treatment early like Carter did. We are thankful that he has not had any symptoms show up and we pray that he will live a happy, healthy and fulfilling life!

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

We need Patient Stories!

BY SHARING YOUR STORY, YOU CAN....

- Spread awareness
- Help others to feel connected
- Share insight beyond our patient community of what it's like to live with homocystinuria
- Provide hope to other families

Ready to share your story?

Send us a message on social media or email us!

info@hcunetworkamerica.org

HCU NETWORK AMERICA

JOIN OUR FUNDRAISING TEAM

We are looking for new community members to join our fundraising team!

Help create, organize and support new and existing fundraising ideas.

These vital funds help support our outreach, programs and research!

To join, email Dbartke@hcunetworkamerica.org

September is Newborn Screening Awareness Month!

Last year we celebrated 60 years of Newborn Screening in the US!

What started as a PKU heel prick blood test developed by Dr. Robert Guthrie (whose own son had PKU!) in 1960, has evolved into the robust system that we know today.

Check out our '***Newborn Screening at a Glance***' infographic to learn more!

Click here to download & share!

NEWBORN SCREENING
at a glance

NEWBORN SCREENING IS A LIFE-SAVER

NBS is a public health program that screens newborns for **serious but treatable** health conditions.

Early detection and **timely treatment** can help prevent serious complications, such as developmental delays, illness, and death.

Approximately **12,000 babies** are diagnosed with a condition through newborn screening in the United States **each year.**

IT CONSISTS OF 3 PARTS
& IS PERFORMED WITHIN THE FIRST 24-48 HOURS OF LIFE

- **blood test**
- **hearing screen**
- **heart screen**

For the blood test, a healthcare provider pricks the baby's heel to collect **a few drops of blood**, which are then placed on a special paper and sent to a lab for testing. The blood test can detect certain genetic, metabolic, hormonal, and functional disorders

IT'S A SCREENING, NOT A DIAGNOSTIC TEST

If a screening test returns an out-of-range result, it indicates a higher risk of a particular condition, but **it does not confirm a diagnosis**. Further diagnostic testing is needed to confirm whether the baby actually has the condition.

NBS IS A FEDERAL PROGRAM, BUT VARIES FROM STATE TO STATE

These differences include the specific **conditions tested**, the **policies for follow-up**, and the **methods** used.

While most states screen for the majority of the RUSP* conditions, some states choose to screen for additional conditions not included in the RUSP. Conversely, some states may not screen for all RUSP conditions due to various factors like cost, infrastructure, or prevalence in the population.

RUSP = *Recommended Uniform Screening Panel; a list of conditions that the U.S. Department of Health and Human Services (HHS) recommends for inclusion in state newborn screening programs. The RUSP serves as a guideline for states to determine which conditions should be part of their newborn screening panels.*

Continued investment in our Newborn Screening programs ensures that **all babies** have **the best possible start in life.**

September is Newborn Screening Awareness Month!

Newborn Screening & Homocystinuria

The approximate number of babies with Classical HCU that are missed at Newborn Screening (however, some reports suggest up to 80% are missed.)

The number of states that screen for classical homocystinuria. Classical HCU was added to the RUSP (Recommended Uniform Screening Panel) in 2007, thanks to the Newborn Screening Saves Lives Act.

The RUSP

(Recommended Uniform Screening Panel)

- The RUSP is a list of disorders that the Secretary of the Department of Health & Human Services *recommends* for states to screen as part of their state universal newborn screening programs.
- Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments.
- Each state ultimately determines what disorders its NBS program will screen for.
- Most states screen for the majority of disorders on the RUSP & some states also screen for additional disorders.
- Massachusetts was the first to adopt HCU newborn screening - in 1968! By 2009, all 50 states were screening for HCU.

https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp

Current Screening Method for classical HCU & its limitations

- Uses methionine level
- Methionine isn't elevated in most HCU patients in 24-48 hours after birth, especially those who are breastfed.
- Each state's newborn screening lab sets its own methionine cut-off to indicate a positive screen.
- A normal methionine level at birth is between 20-30.
- If a lab sets its methionine cut-off too high it will miss patients.

"Elliott's classical HCU was missed at newborn screening because his methionine level was not yet elevated enough to receive a positive screen. His level was 44 and to receive a positive screen in South Carolina, it must be 65."

-Liz Carter, mom of Elliott, diagnosed with classical HCU at age 2 1/2

Cobalamin Disorders & Severe MTHFR

Combined Cobalamin Disorders are a part of the RUSP Secondary Conditions. This means they are picked up as a result of a different disorder, typcally Methylmalonic acidemia. These condtions are Cobalamin C, F, J, K, X and TC II.

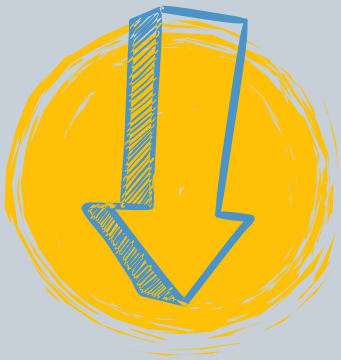
Some states set a low methionine cut off and these will flag the conditions above, but also flag Isolated Cobalamin Disorders, Cbl D, E, G & Severe MTHFR.

New developments give hope for the future

In 2023 the CDC came up with a 1st tier multi-plex approach that would allow total homocysteine to be the primary marker in screening for Homocystinuria.
The goal? A more accurate screen, and identifying all babies with homocystinuria at birth.

Learn more:
https://hcunetworkamerica.org/newborn-screening/
https://academic.oup.com/clinchem/article/69/5/470/7068836?login=false

HCU NETWORK AMERICA



Click here to download & share!

September is Newborn Screening Awareness Month!

Congratulations to the CDC Newborn Screening and Molecular Biology Branch!

TThe CDC Newborn Screening and Molecular Biology Branch team's 2023 manuscript, titled "***Multiplexing Homocysteine into First-Tier Newborn Screening Mass Spectrometry Assays Using Selective Thiol Derivatization,***" received the prestigious "Visibility Award" from the Association for Diagnostics & Laboratory Medicine (ADLM). This award was given for achieving the highest Altmetric score among all original research articles published in 2023, highlighting the manuscript's global impact and widespread attention across social media, news outlets, policy documents, and other platforms.

Clinical Chemistry 69:5
470–481 (2023)

Proteomics and Protein Markers

Multiplexing Homocysteine into First-Tier Newborn Screening Mass Spectrometry Assays Using Selective Thiol Derivatization

C. Austin Pickens,* Elya Courtney,* Samantha L. Isenberg,* Carla Cuthbert,* and Konstantinos Petritis*

BACKGROUND: Classical homocystinuria (HCU) results from deficient cystathionine β-synthase activity, causing elevated levels of Met and homocysteine (Hcy). Newborn screening (NBS) aims to identify HCU in pre-symptomatic newborns by assessing Met concentrations in first-tier screening. However, unlike Hcy, Met testing leads to a high number of false-positive and -negative results. Therefore, screening for Hcy directly in first-tier screening would be a better biomarker for use in NBS.

METHODS: Dried blood spot (DBS) quality control and residual clinical specimens were used in analyses. Several reducing and maleimide reagents were investigated to aid in quantification of total Hcy (tHcy). The assay which was developed and validated was performed by flow injection analysis–tandem mass spectrometry (FIA-MS/MS).

RESULTS: Interferents of tHcy measurement were identified, so selective derivatization of Hcy was employed. Using N-ethylmaleimide (NEM) to selectively derivatize Hcy allowed interferent-free quantification of tHcy by FIA-MS/MS in first-tier NBS. The combination of tris(2-carboxyethyl)phosphine (TCEP) and NEM yielded significantly less matrix effects compared to dithiothreitol (DTT) and NEM. Analysis of clinical specimens demonstrated that the method could distinguish between HCU-positive, presumptive normal newborns, and newborns receiving total parenteral nutrition.

CONCLUSIONS: Here we present the first known validated method capable of screening tHcy in DBS during FIA-MS/S first-tier NBS.

Introduction

Homocystinuria (HCU) results from the inability to convert homocysteine (Hcy) to cystathionine due to enzyme (1) or vitamin B12 deficiencies (2), causing an elevation of Hcy in blood and urine (3). Classical HCU [OMIM #236200] results from deficient activity in the transsulfuration enzyme cystathionine β-synthase (CBS), a vitamin B_6-dependent enzyme, causing elevated levels of Met and Hcy. The global rate of HCU is 1 in 200 000 to 335,000, and its frequency varies based on geographic location, (1 in 65 000 in Ireland; 1 in 17 800 in Germany; and 1 in 1800 in Qatar (4)). Complications of untreated HCU include ocular issues (i.e., ectopia lentis), skeletal abnormalities, dysfunction and abnormalities of the vascular system (e.g., thromboembolism and vascular occlusions), developmental and intellectual disabilities, failure to thrive, and symptoms similar to Marfan syndrome (1, 5). Decreased morbidity and improved outcomes are associated with early interventions in HCU-positive (HCU+) individuals (6–8), with treatments ranging from vitamin B_6 and/or betaine supplementation to Met-controlled and/or low-protein diets (1, 5). Identification of pre-symptomatic HCU+ individuals is possible through newborn screening (NBS), the routine screening for dozens of inborn errors of metabolism shortly after birth using dried blood spot (DBS) analyses. HCU was added to the US Recommended Uniform Screening Panel (RUSP) in 2006 (9).

Current first-tier NBS methods for HCU utilize Met as a biomarker. In some NBS laboratories, presumptive HCU+ are reflexed to second-tier screening to assess Hcy concentrations. However, Met concentrations are often not elevated enough 24 to 48 h after birth

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Disclaimer: The findings and conclusions in this study are those of the authors and do not necessarily represent the views of the US Department of Health and Human Services, or the US Centers for Disease Control and Prevention. Use of trade names and commercial sources is for identification only and does not constitute endorsement by the US Department of Health and Human Services, or the US Centers for Disease Control and Prevention.
Received August 8, 2022; accepted January 9, 2023.
https://doi.org/10.1093/clinchem/hvad007

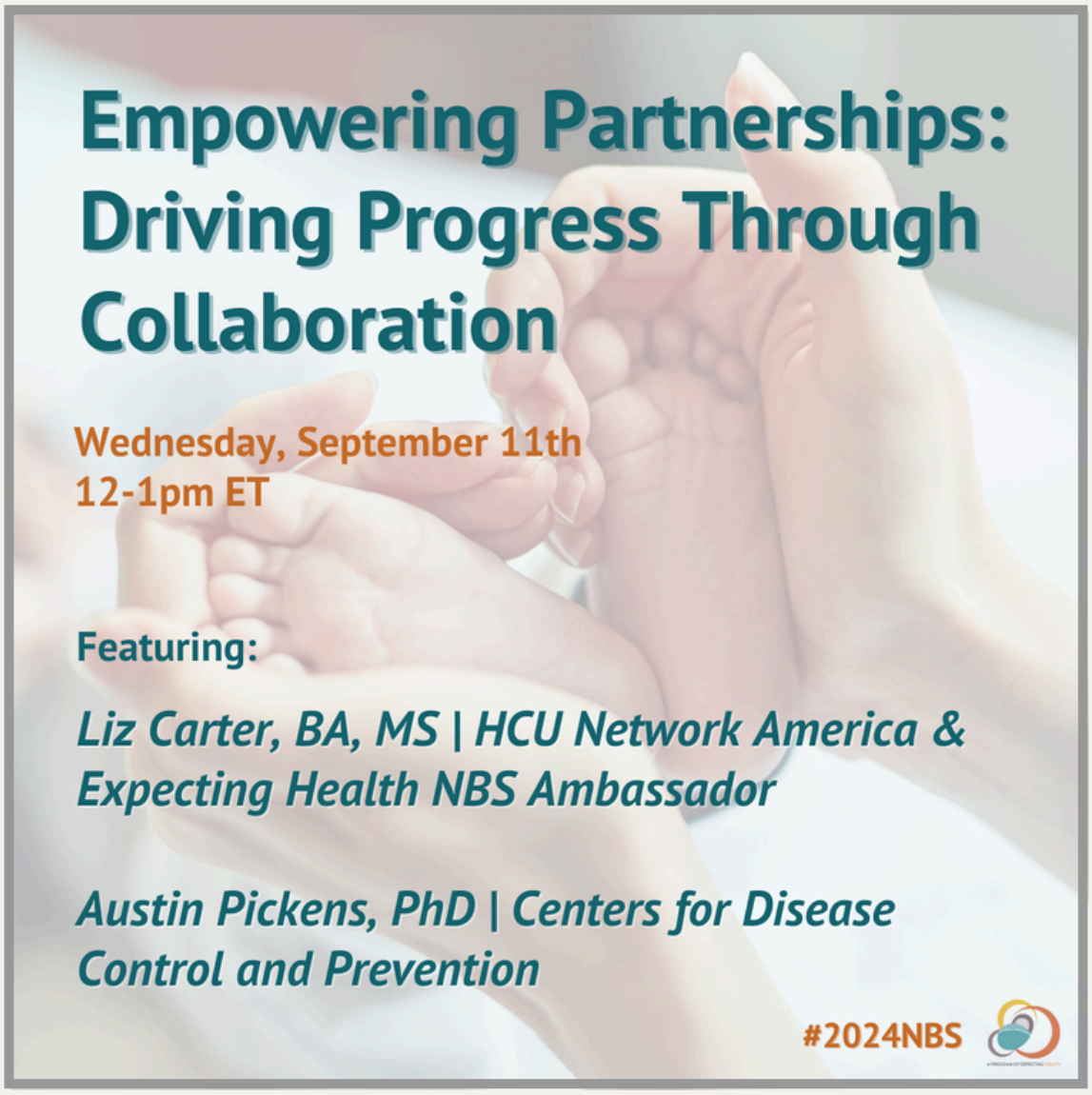
Published by Oxford University Press on behalf of American Association for Clinical Chemistry 2023.
This work is written by (a) US Government employee(s) and is in the public domain in the US.

470

Read the manuscript here

Pictured left to right:
Liz Carter (HCU Network America), **Elya Courtney** (collaborator), **Danae' Bartke** (HCU Network America), **Samantha Isenberg** (collaborator), **Austin Pickens** (project lead), **Kostas Petritis** (principal investigator [PI]).
Not pictured: **Carla Cuthbert** (branch chief)

Newborn Screening Awareness Month



In this webinar, Liz Carter & Austin Pickens will discuss how the collaborative partnership between HCU Network America and the CDC has driven progress for newborn screening, and what it means for the HCU Community & others.

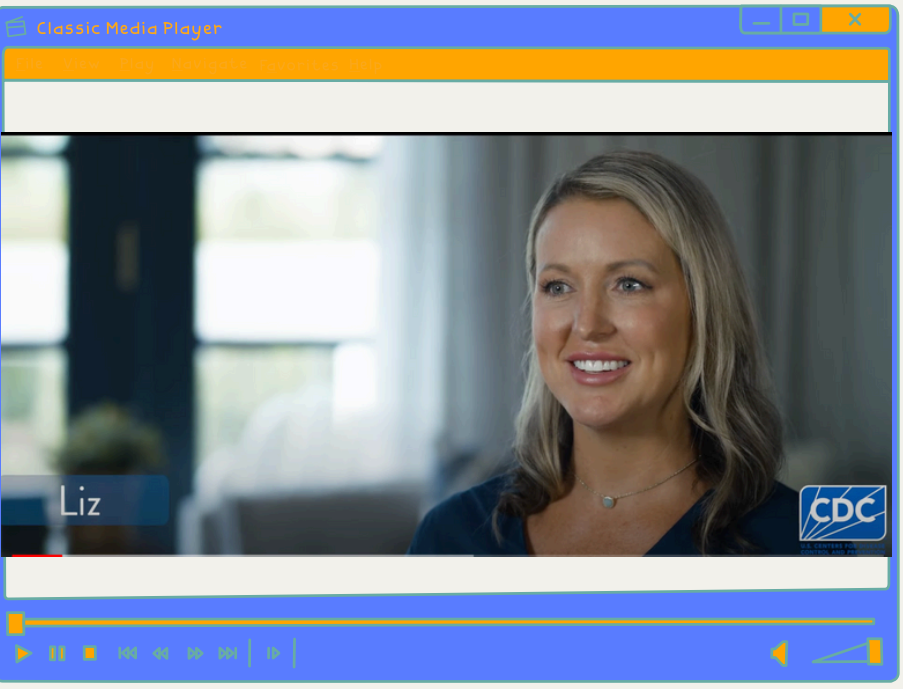
Register here!

Newborn Screening & HCU

History of Homocystinuria Newborn Screening with HCU Pioneer, Dr. Harvey Levy

HCU Network America Vice President Mark Lewis interviews the world-renowned Dr. Harvey Levy about his involvement and the history of newborn screening for the Homocystinurias.

Click to watch

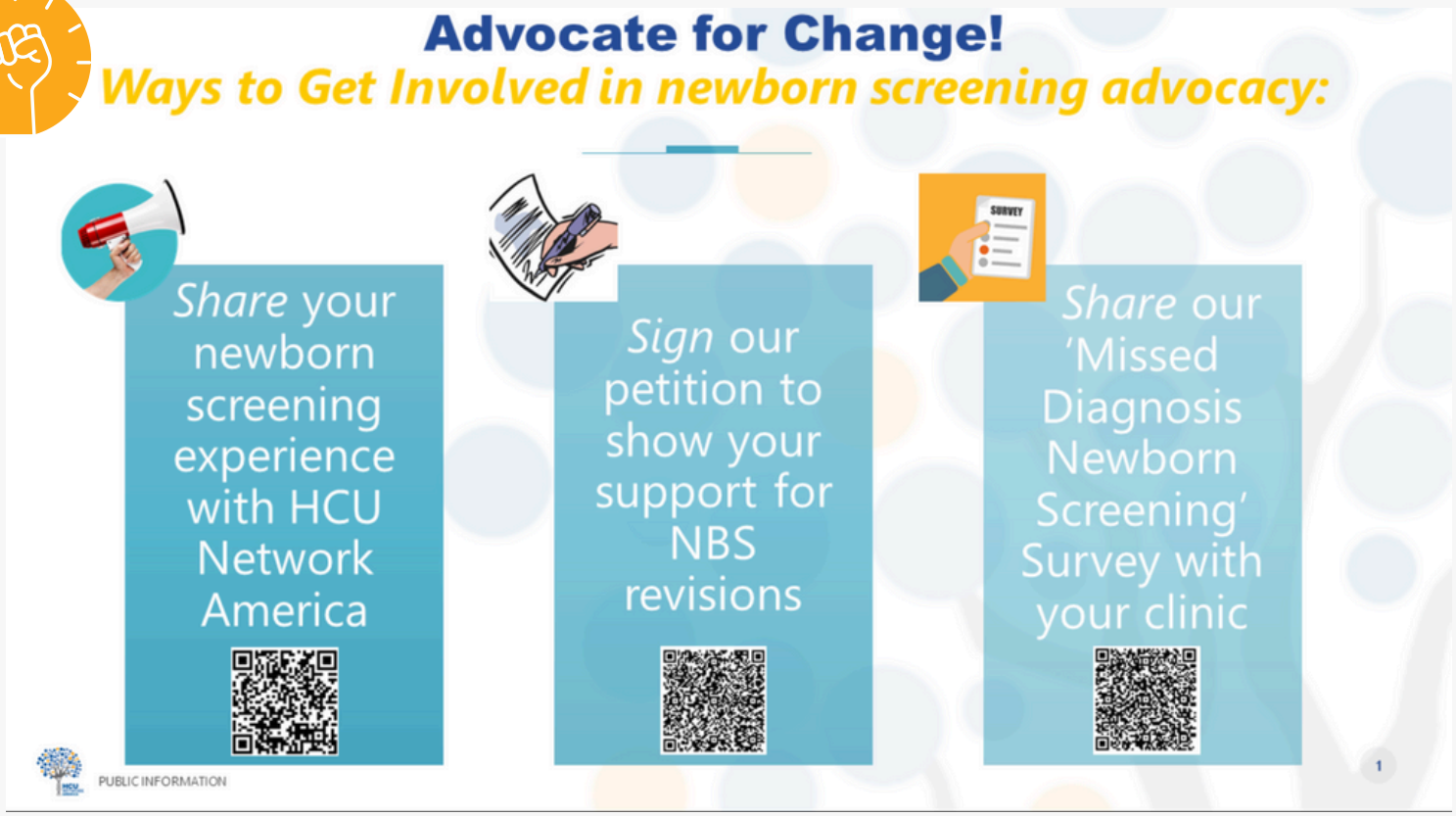


CDC Newborn Screening: "Homeward Bound"

Liz, the mom of a child with homocystinuria who was missed by Newborn Screening, shares her family's story.

Click to watch

Newborn Screening Awareness Month



Advocacy Tips, Tricks & Ideas

- Share one (or both!) of our infographics, [along with something about your newborn screening story if you'd like], to social media during the month of September!
- Don't want to create your own posts for social media? That's ok! Keep an eye on HCU Network America's posts and share them!
- Check out Baby's First Test NBS Awareness Social Media Toolkit! It's got some ready-made posts and graphics that you can use!
- Use the hashtags: **#NewbornScreening** & **#2024NBS** to give your posts more visibility!
- Not a social media person? No worries! Make it a point to tell someone (*a friend, teacher, coworker, doctor, or even the barista at your coffee shop*!) that September is Newborn Screening Awareness Month and share a little bit with them about your (or your child's) newborn screening story.

UPCOMING EVENTS

REGISTER

On your marks...Get set....GO!

Our Race for Research has officially kicked off!

It's still not too late to join the fun!

You can sign up (with as little as $5 to participate!) here!

Don't forget to log your miles!

Racers can sync their page to Strava OR visit your team's race page to log miles manually!

THANK YOU to our amazing sponsors for helping to make this year's Race for Research a success!

eTon PHARMACEUTICALS

Learn more about Eton Pharmaceuticals

RECORDATI RARE DISEASES
Focused on the Few®

Learn more about Recordati Rare Diseases

Meet our Teams!

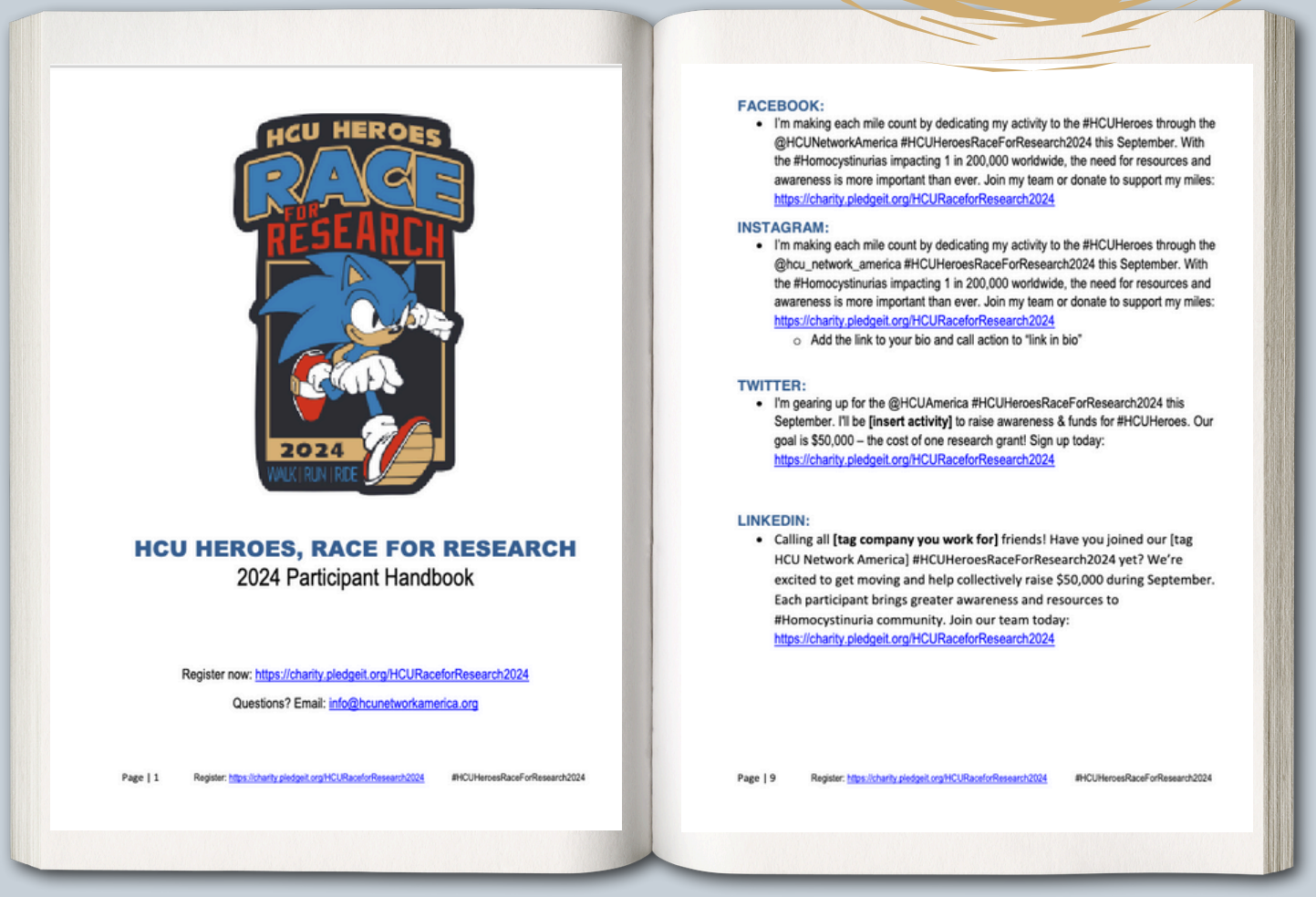
Click to read each team's story & donate!

 Amy's Army	Click here
 Brooke's Blazers	Click here
 Carson's Cure Chasers	Click here
 Carter Crew 4 HCU	Click here
 CblC Brazil	Click here
 Cure for Casey	Click here
 Ellie's Entourage	Click here
 Grayson's Gang	Click here
 "Hunt" for research	Click here
 March for Mariella	Click here
 Masen's Mad Dawgs	Click here
 Miles for Marcus	Click here
 Miles for Marley	Click here
 Sarah's Sidekicks	Click here
 Sonic's Sprinters	Click here
 Team Anniston	Click here
 Team Butter Stick	Click here
 Team Dayton	Click here
 Team Recordati	Click here
 Team Will for HCU	Click here
 Teo's Trailblazers	Click here
 The Bartke Ruff Ruffs	Click here
 Traverians	Click here

Follow the leaderboard throughout the month!

https://charity.pledgeit.org/HCURaceforResearch2024

To date, our Race for Research has funded 7 research grants for HCU!



HCU HEROES RACE FOR RESEARCH 2024 WALK | RUN | RIDE

HCU HEROES, RACE FOR RESEARCH
2024 Participant Handbook

Register now: https://charity.pledgeit.org/HCURaceforResearch2024

Questions? Email: info@hcunetworkamerica.org

Page | 1 Register: https://charity.pledgeit.org/HCURaceforResearch2024 #HCUHeroesRaceForResearch2024

FACEBOOK:

- I'm making each mile count by dedicating my activity to the #HCUHeroes through the @HCUNetworkAmerica #HCUHeroesRaceForResearch2024 this September. With the #Homocystinurias impacting 1 in 200,000 worldwide, the need for resources and awareness is more important than ever. Join my team or donate to support my miles: https://charity.pledgeit.org/HCURaceforResearch2024

INSTAGRAM:

- I'm making each mile count by dedicating my activity to the #HCUHeroes through the @hcu_network_america #HCUHeroesRaceForResearch2024 this September. With the #Homocystinurias impacting 1 in 200,000 worldwide, the need for resources and awareness is more important than ever. Join my team or donate to support my miles: https://charity.pledgeit.org/HCURaceforResearch2024
  - Add the link to your bio and call action to "link in bio"

TWITTER:

- I'm gearing up for the @HCUAmerica #HCUHeroesRaceForResearch2024 this September. I'll be **[insert activity]** to raise awareness & funds for #HCUHeroes. Our goal is $50,000 – the cost of one research grant! Sign up today: https://charity.pledgeit.org/HCURaceforResearch2024

LINKEDIN:

- Calling all **[tag company you work for]** friends! Have you joined our [tag HCU Network America] #HCUHeroesRaceForResearch2024 yet? We're excited to get moving and help collectively raise $50,000 during September. Each participant brings greater awareness and resources to #Homocystinuria community. Join our team today: https://charity.pledgeit.org/HCURaceforResearch2024

Page | 9 Register: https://charity.pledgeit.org/HCURaceforResearch2024 #HCUHeroesRaceForResearch2024

Our Race Participant handbook has lots of good info & tips for a successful race & fundraiser!

Download it here

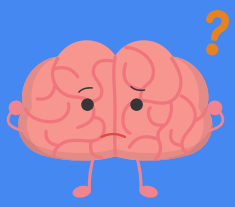
WHAT IS HOMOCYSTINURIA?

Homocystinuria denotes an elevation of homocysteine or homocystine in the urine.

Elevations in the blood may indicate an inherited form, but is not always the case.

Homocystinuria affects
1 in 200,00 people worldwide.

HOMOCYSTINURIA, THERE IS MORE THAN ONE?



B6 RESPONSIVE CLASSICAL HOMOCYSTINURIA

B6 NON-RESPONSIVE CLASSICAL HOMOCYSTINURIA

SEVERE MTHFR

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN C

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN D

HOMOCYSTINURIA, COBALAMIN D

HOMOCYSTINURIA, COBALAMIN E

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN F

HOMOCYSTINURIA, COBALAMIN G

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN J

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN K

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN X

Classical homocystinuria is screened for in all 50 states, and most states screen for the various forms of methylmalonic acidemia with homocystinuria, however, it's estimated that at least 50 % of cases are missed! Without a diagnosis or early intervention, all homocystinurias can be fatal!

THERAPY?

The last approved therapy for the homocystinurias was approved in 1996.

Even with this therapy, patients remain at risk with high levels of homocysteine.

HELP ACCELERATE RESEARCH

7
Research Grants Funded

GRANT

$268,000
Total research grants awarded and counting

MEET OUR NEW MEDICAL ADVISOR!

MEET OUR MEDICAL ADVISOR

Josh Baker,
DO, FAAP, FACMG

Dr. Josh Baker is the Director of Inborn Errors of Metabolism in the Division of Genetics, Genomics and Metabolism at Lurie Children's Hospital. He specializes in diagnosis, treatment, and management of IEMs. He is also the Director of Newborn Screening at Lurie Children's Hospital and Chair of the Illinois Newborn Screening Committee. He participates in several clinical trials as principal investigator in novel therapies to treat IEMs. Current independent research includes investigating the social determinants of heath and impact on care of metabolic disease, the diagnosis of Cobalamin disorders in Illinois, and treatment impact of cobalamin disorders using high dose hydroxocobalamin.

HCU Champions

INTRODUCING

HCU CHAMPIONS

HCU Champions is a new program designed to help increase awareness and advocacy for homocystinuria (HCU).

HCU Champions will share their journeys and experiences of living with HCU to help inspire, motivate, and empower individuals to connect and support one another in their communities and beyond.

Get involved!

Incase you missed it...

The HC&U Podcast is back!!!

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben & Lindsey.

Meet your hosts!

Welcome to the HC&U Podcast! We are Ben and Lindsey, your hosts. We are so excited to be starting this as extra resources for the Homocystinuria community. We hope you like our content!

To Listen:

https://hcunetworkamerica.org/hcu-podcast/

or click below on your favorite option!

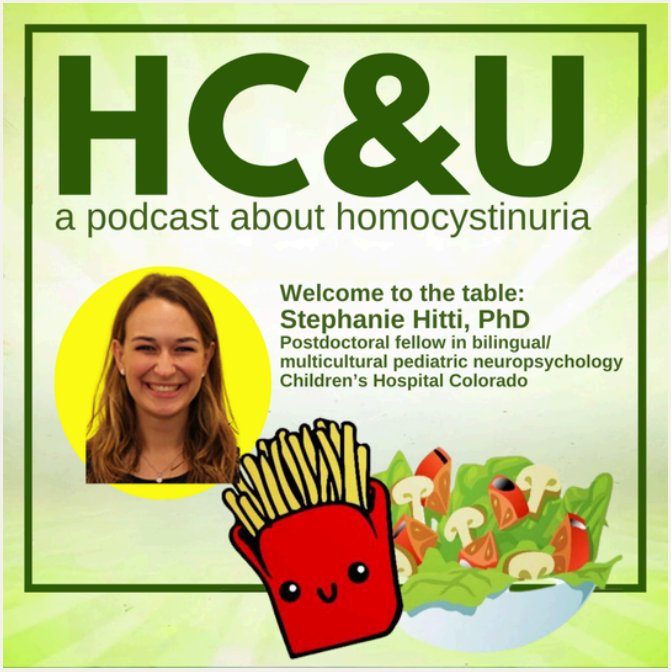
LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

The latest episode

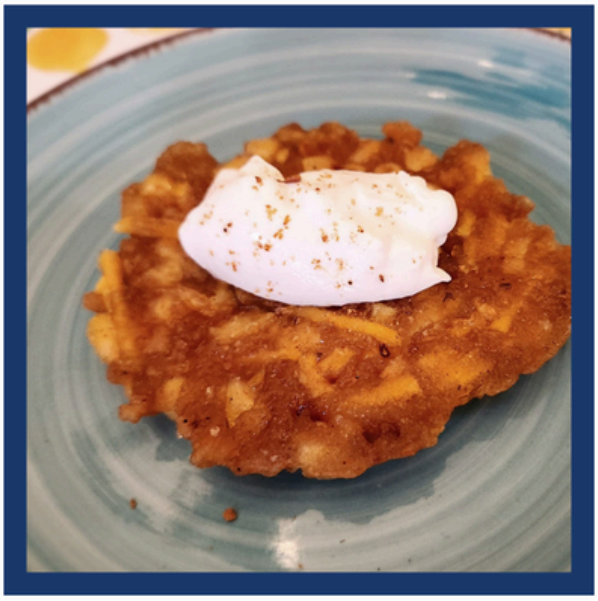


This time, Ben welcomes Dr. Stephanie Hitti to the table!

Specializing in brain development, Stephanie uses this training to evaluate & help manage children with health conditions that affect the brain.

Join Stephanie & Ben as they discuss some of the neurocognitive risk factors in children with the homocystinurias, as well as navigating the world of school accommodations! (Ben is a high school teacher and an adult living with HCU, so he'll have plenty of insight to add!)

Corn Fritters



Makes 4 Fritters | 1.1 gram protein per Fritter

Ingredients:

- 84 g Corn Kernels, Thawed if frozen or drained if canned
- 1 TBSP Scallions (green parts) chopped
- 1 clove(s) Garlic, minced
- 3/4 c Wheat Starch
- 2 tsp Taco Seasoning
- 1/4 c Vegetable Broth
- 1 oz. Violife Just Like Cheddar Shreds

Directions:

1. In a medium bowl, add the corn, scallions, garlic, and taco seasoning. Gently combine. Add the wheat starch and the vegetable broth and cheese. Whisk together to make a batter. Set aside.

2. Heat vegetable oil in a medium skillet over medium heat. You want about 1/2 inch of oil in the skillet. Using a 1/4 cup measuring cup, pour the batter into the hot oil. Allow to cook until the bottom is nicely browned, about 2-3 minutes. Gently flip and allow the other side to cook until browned, about 2-3 minutes. Remove from pan and place on a paper towel lined place to absorb the excess grease. Continue cooking the rest of the batter the same way. Serve hot with your choice of toppings.

Note: You can use water in place of the broth. You can use any seasoning you desire. I actually used bacon seasoning when I created this recipe. You can play around with other veggies in this recipe if needing to adjust protein content.

GO BLUE FOR HCU

October is HCU Awareness Month

Click here to order your gear!

HCU Awareness

All proceeds from the sale of our gear benefit HCU Network America!

It's HCU Awareness Month –
LET'S TALK FUNDRAISER SETUP!

There are many ways that you can participate in HCU Awareness Month.

One way is by hosting a fundraiser!

Not only do fundraisers raise vital funds to help us support the Homocystinuria community, they spread awareness of the Homocystinurias and the challenges of living with the conditions.

Not sure how to get started?
Let our Fundraising committee help!

Email
info@HCUnetworkamerica.org

Click here for a list of additional HCU Awareness and Fundraising Event Ideas!

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

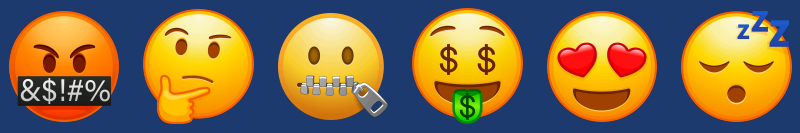
POWERED BY RARE X

What is the Vineland-3?

The Vineland-3 is designed to help you efficiently identify your child's strengths and weaknesses and can help qualify them for services if needed.

- The traditional Vineland-3 has 3 parts.
  - Interview Form
  - Parent/Caregiver Form
  - Teacher Form
- The Rare-X Vineland 3 survey solely relies on the input of parents/caregivers.
  - The form includes 3 domain areas, with 9 subdomains.

Communication	Daily Living Skills	Socialization	Motor Skills (Optional)
• Receptive • Expressive • Written	• Personal • Domestic • Community	• Interpersonal • Play/Leisure • Coping Skills	• Fine Motor • Gross motor



Complete the Vineland 3 Assessment at https://homocystinuria.rare-x.org

CANPCU+ CANPKU+ 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

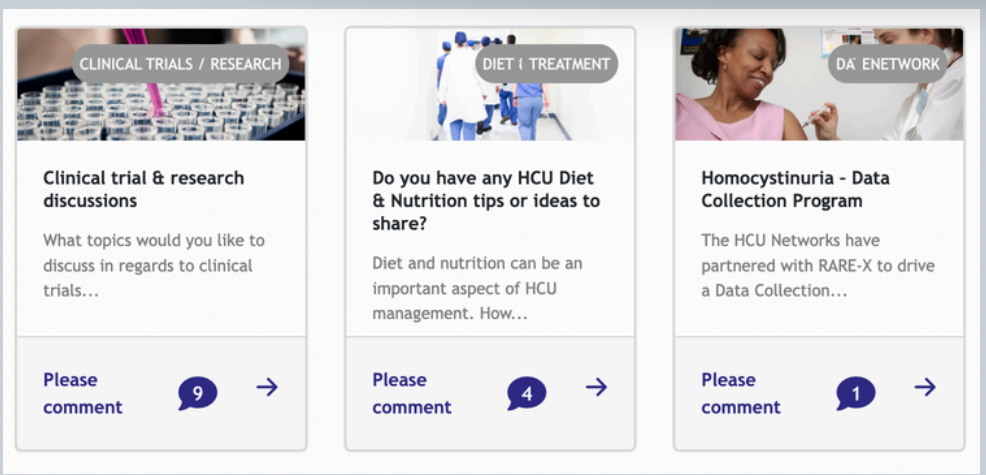
TRAVERE THERAPEUTICS

JOIN THE HCU E-NETWORK!

Welcome to the HCU eNetwork

Powered by HCU Network America & HCU Network Australia, we aim to utilize this platform to connect with HCU patients and carers worldwide and gather your input on key topics in relation to HCU diagnosis, management and treatment.

Questions and activities will be updated on the platform throughout the year, so please check back regularly and look out for email communications that will be sent out notifying you when new topics are posted.



HCU NETWORK AMERICA

HCU Network Australia

Join the conversation!

https://hcuenetwork.org/

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*!

UPDATE

Now includes funding assistance for medical formula AND low protein foods!

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

MEDICAL ASSISTANCE

What is the purpose of this program?

Having a rare disease is difficult. Adding in the complex care required to treat or manage that disease and figuring out how to pay for it makes a rare diagnosis even harder.

NORD's Classical HCU Medical Assistance Program offers eligible individuals diagnosed with Classical Homocystinuria financial support to pay for the low protein foods and physician prescribed medical formulas necessary in managing this HCU diagnosis.

NORD's HCU Medical Assistance Program opened thanks to a generous donation from the HCU Network America.

Who is eligible to apply?

This program is designed to help patients who:

- Have a diagnosis of Classical Homocystinuria.
- Are a United States citizen or U.S. resident of six (6) months or greater with evidence of residency such as a utility bill showing the patient's name and address.
- Meet the program's financial eligibility criteria.

What is the application process?

Patients may be referred to the program by their health care provider, their case managers, or they may self-refer.

A NORD Patient Services Representative will guide the applicant through the application process and verify eligibility for inclusion in the HCU Program.

Awards are based on meeting eligibility criteria, funding availability, and are made on a first-come, first serve basis.

NORD is Here for You

NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.

NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

Monday-Thursday 8:30am – 7:00pm ET
Friday 8:30 am – 6:00pm ET

203-616-4327

203-635-4163

hcu@rarediseases.org

US MAIL to: NORD
Attention: HCU Program
55 Kenosia Avenue
Danbury, CT 06810

What assistance does NORD provide?

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods and physician prescribed medical formulas. Individuals approved for assistance in this program will be issued a PEX card. The PEX card is a prepaid expense card to be used for the purchase of low protein foods or physician prescribed medical formulas only.
- *Upon receipt of the card, the cardholder will contact NORD to request card activation. The card will be funded based on program award caps set for the program (this cap will be discussed with individuals upon enrollment in the program..*
  - *> It is necessary for the individual to submit receipts on a monthly basis evidencing card utilization for the purchase of low protein foods or medical formulas for the previous month.*
  - *> Funds will not be added to the card until the previous month's receipts have been received by NORD.*
  - *> The card may only be utilized for the purchase of low protein foods up to the monthly program limit.*

Once a patient is accepted into the assistance program(s) how long are they eligible?

Awards are issued for a calendar year.

Patients are encouraged to reapply annually if continued assistance is needed.

What happens if an applicant does not meet the criteria of the Electronic Income Verification?

The NORD Patient Services Representative will offer to e-mail, fax, or mail the brief program application and disclosure forms to the patient. The applicant may then complete the application, sign the disclosure form, provide the appropriate financial documentation to verify financial need, and return them via fax, email, or USPS mail.

How does NORD demonstrate compliance with regulations required of charities?

- NORD independently designs its patient assistance programs based on the needs of specific patient communities.
- No pharmaceutical company or donor controls or influences our programs.
- Our patient assistance decisions are based on consistently applied financial eligibility criteria and diagnosis only.
- Patients have their choice of health care provider, treatment and treatment location, and can make changes at any time.
- Patients' privacy and well-being are priorities at NORD. We do not share or provide patient names or data with donors, nor do we disclose or identify donors to patients. Patients are able to make the choices that are best for them because NORD's assistance covers all FDA-approved products available for a diagnosis. Our programs also help with more than medication: patients can use their funds to pay for other physician prescribed services related to their diagnosis, such as laboratory and diagnostic testing, physical and occupational therapy, durable medical and adaptive equipment, and travel to medical appointments.

NORD®

rarediseases.org

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