The HCU Herald

October is HCU Awareness Month!

Feauturing...

HCU Hero

Zoraiz & Areeba from Virginia

October 2025

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

HCU HERO:
ZORAIZ & AREEBA FROM VIRGINIA

Zoraiz was born in Virginia following an uncomplicated full-term pregnancy and c-section birth. The only notable complication was having jaundice but lots of kids have that after birth, so we didn't think anything of it. She was drinking fine at first for a few days but stopped drinking her milk on and off, so I took her to the pediatrician. Her jaundice was bad, so she was taken back to the hospital for treatment for a few days.

When we went home, she was still struggling to drink as her suction was not strong. She was also sleeping a lot more than usual. After a few weeks, I took her to the pediatrician

again and told them I thought something was going on. They dismissed me and said I was just a young mom and didn't know how to raise a child. She was my first child, but I knew that something didn't seem right. At 45 days old I took her back to the pediatrician because when I woke up in the morning, I lifted her arm, and it just flopped back down. I checked her nose, and it seemed like she was breathing but very shallow; then I noticed her lips were also bluish. The pediatrician told us to go to the hospital, so my dad came to get me, and we took her. At first, they thought she had pneumonia and inserted a breathing tube. She was there for two weeks while they were figuring out what was going on. They did an MRI of her brain and discovered atrophy and big ventricles compared to a normal child, but they still didn't know what was causing it.

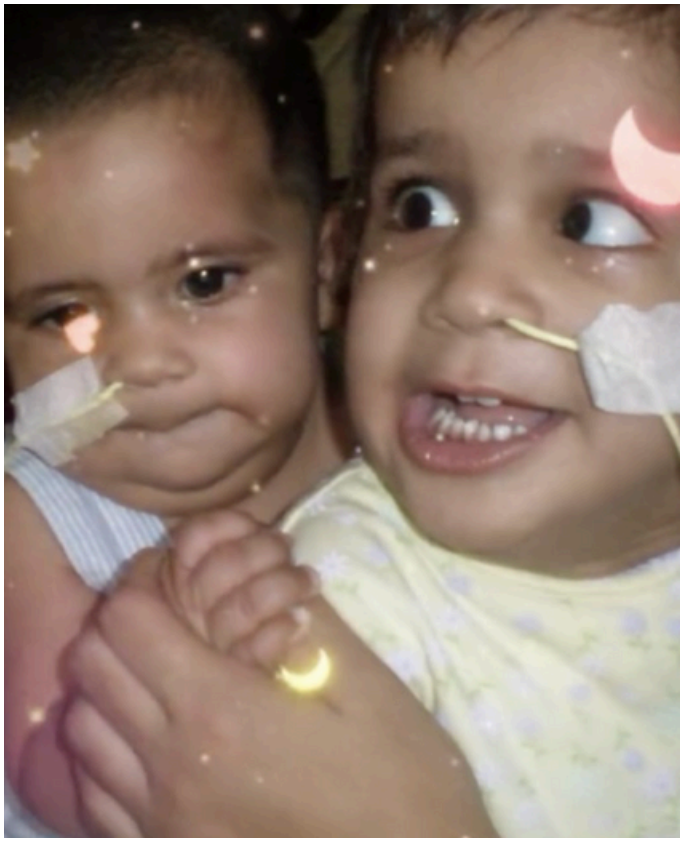
HCU HERO:
ZORAIZ & AREEBA FROM VIRGINIA

The hospital contacted Children's in D.C for assistance. She started having seizures at that time as well, so they decided to transfer her. When we arrived at the children's hospital we met with the genetic team where they asked us all kinds of questions and ran blood tests. She was still on the ventilator and hooked up to electrodes to monitor her seizure activity. It was a very difficult time because we were over an hour bus ride from the hospital. We finally met with the genetic doctors, and they told us she had homocystinuria caused by MTHFR. I had no idea what that was.They explained that it is very rare and only 50 or so people in the world had this disorder.

They showed us the MRI and how her brain was shrinking, and her ventricles were getting bigger. They told us it was like a roller coaster that's going down the hill and they are trying to catch it before it reaches the bottom.

HCU HERO:
ZORAIZ & AREEBA FROM VIRGINIA

That was really hard to hear about your child. We stated to give her medication through an NG tube, and she started to show slow progress. I remember signing this waiver to take her off the ventilator because she was on it for two months. It stated that she could die if we took her off and that was hard. They asked me if I wanted to send her to a facility to take care of her, but I wanted her home with me. She stayed in the NICU while I learned to



place an NG tube and administer her medication. We finally came home with lots of equipment. I never knew so many things existed to help my daughter stay alive. A VP shunt was eventually placed as she had hydrocephalus.

They told me her prognosis was around six months and that she could pass away in her sleep or have a stroke. I understood what could happen but knew she would be ok as long as she was with me.

About a year later we had our second daughter, Areeba. My geneticist from Children's gave me a letter to instruct the doctors to run blood work as soon as she was born. Unfortunately, she had the same gene mutation as her homocysteine level at birth was around 300. They quickly transferred her to the NICU, and she was there for a few weeks while we began medication and lowered her homocysteine levels. Just like that I came home and had two kids on NG tubes taking the same medications. Areeba did much better, although did still have some delays.

HCU HERO:
ZORAIZ & AREEBA FROM VIRGINIA

She started crawling at 8 months and walking when she was 2. She received OT, PT and early intervention. She was delayed but not as significantly as Zoriaz. Zoraiz started walking at the age of 12 because we never gave up. She also received Speech, OT and PT services.

Zoriaz is now 19 and Areeba is 18. Areeba still doesn't talk but she has a talker and had an IEP in school. She is developmentally around the age of a 6- or 7-year-old. I see the difference between the two sisters and know that Zoriaz struggles more than Areeba because it took the doctors so long to figure out what was wrong.

HCU HERO:
ZORAIZ & AREEBA FROM VIRGINIA

I am so grateful for the team of doctors at Children's who have been so supportive and helpful with my kids on this journey. It's been difficult this past year as Zoraiz is in a lot of pain and is bedridden.

We have been in and out of the hospital every 2-3 weeks for the past year. I just want her to be comfortable. It is amazing that she has fought for 19 years and exceeded every expectation. I couldn't ask for more from her.

She's a fighter and she taught me to be who I became. I never thought I would be this strong.

Share Your Story

With Us!

To Join or Learn More:
Email
info@hcunetworkamerica.org

JOIN OUR FUNDRAISING TEAM

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

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

To Join or Learn More:
Email
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UPCOMING EVENTS

HCU AWARENESS MONTH

VIRTUAL MEET UP

CLICK HERE!

SUNDAY 12 OCTOBER

4 PM CT/5 ET/ 3 MT

UPCOMING EVENTS

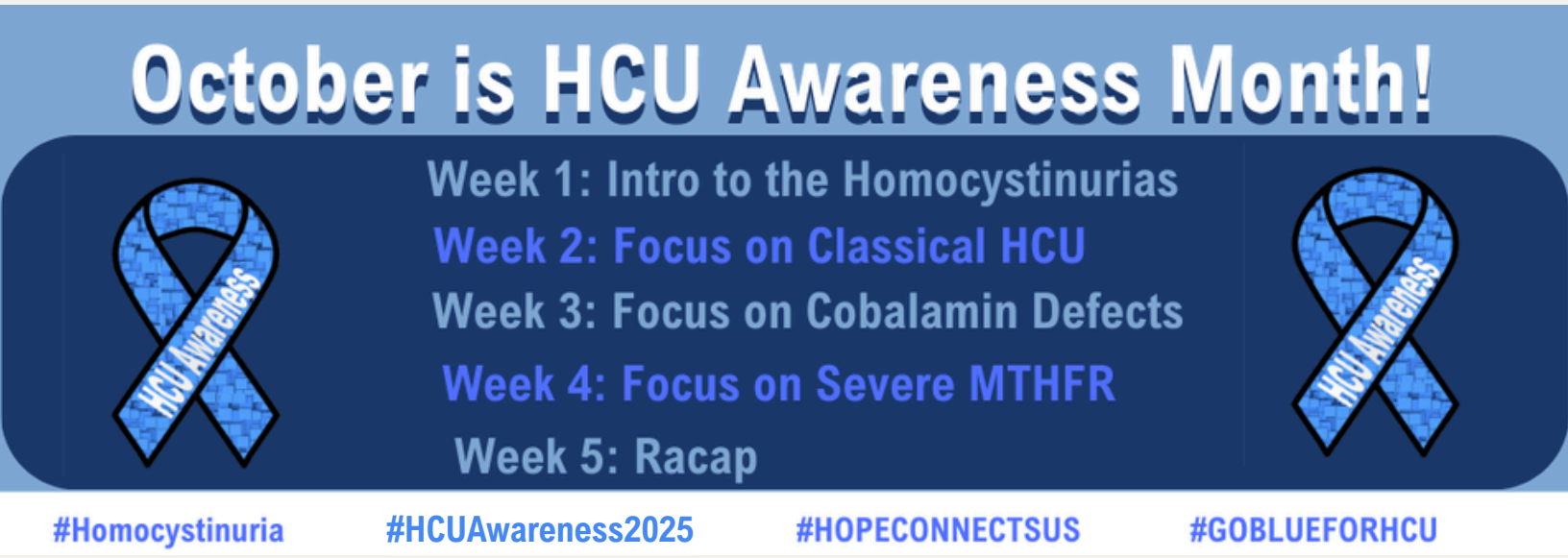
WEAR YOUR AWARENESS

October is HCU Awareness Month!
Get your Gear NOW!

Click Here


STICK WITH AWARENESS
Get your limited edition
HCU Stickers and Magnets
Click Here

HCU AWARENESS MONTH: LEVERAGING SOCIAL MEDIA!



One of the best ways to reach a wide audience to spread awareness for HCU is through social media!

How can you participate?

- Share HCU Network America's posts
- Create your own original posts (see our *Social Media Choiceboard* on the next page for ideas!)
- Share/comment on other posts from those in the community
- Start a social media fundraiser for HCU Awareness Month

Click to follow us!

HCU AWARENESS MONTH SOCIAL MEDIA CHOICEBOARD

How to use:

- Click **here** to download your copy with clickable links.
- Throughout the month, simply choose which items you'd like to post!
- Tag HCU Network America in your posts!

October is #HCUAwareness month! Show your support for the #HCU community by changing your profile to the awareness ribbon! **Awareness ribbon**	Start a fundraiser & share it out on your social media! Platform ideas: • Facebook or Instagram Fundraising • Gofundme (example) • Give Lively [Visit our fundraising page for more ideas]	#FacesofHCU: I am 1 in 200,000 people living with #HCU. *insert your photo*	Share a picture of you & an HCU Buddy, or tag a friend who is a great support. *insert your photo*
Share your/your child's patient story from our website (if you have one there). If not, share a little bit about your diagnosis. [Find the link to your story: https://hcunetworkamerica.org/patient-stories/]	Amplify another fundraiser in the community! Share out another person's fundraiser.	Share a picture of you/your family in an HCU tshirt/gear	Share a picture or video of your daily medications + formula (for those who take formula)
Share a favorite quote and how it connects to your HCU story or your outlook on living with HCU.	#HCUHero: Share/feature someone in the HCU Community who inspires you	#Hope4HCU: Share what gives you encouragement & hope	Travel with #HCU! Share a picture of your 'extra packing' to accommodate life with HCU, or your favorite travel hack!
Share a video from our Youtube Channel & why it resonates with you.	Listen to an episode of HC&U Podcast & share our your key takeaways or a quote that resonated with you	For those on a low-protein diet, share your daily allotment and a photo of a meal	Post of a picture of you/your child doing your favorite activity.
Share a picture of you from one of our conferences and tell why it was a meaningful experience for you.	For those on low-protein diet: Share your favorite low-protein meal or snack along with how many grams of protein per serving it contains.	For those on low-protein diet: Share your favorite restaurant with low-protein options & what you order there	Share something that you want people to know about HCU
Share something interesting / fact(s) from our Classical HCU Toolkit	Share something interesting / fact(s) from our Cobalamin Disorders Toolkit	Share out our MTHFR: Could it be homocystinuria? One-pager Sample post text: MTHFR, Severe MTHFR? What's the difference? MTHFR is one of the most confusing of the homocystinurias, as the common mutations are nothing like the severe form! Read to find out more.	For those with vision issues: share how this has impacted you.
Appreciate your clinic/team! Give a shout out to your clinic, team, or a specific provider who has been a champion for you in your HCU journey. If you can, tag them/the clinic in your post!	What is the most helpful piece of advice you've gotten? Share it out!	What does #community mean to you? How has finding a community of others who live with HCU been important to you?	#HopeConnectsUs: Share what your hopes are for a future therapy/treatment

IT'S HCU AWARENESS MONTH- LET'S TALK FUNDRAISER SET UP!

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!

ZOIA Pharma is proud to offer a wide range of medical formulas, low protein foods and enteral nutrition from leading manufacturers to support your nutritional management needs.

Nourishing **your unique nutritional journey**

As your one-stop for metabolic home medical and supplies, we are pleased to help serve the Homocystinuria community. Learn how ZOIA Pharma is dedicated to serving patients with this inherited metabolic disorder.

To discover how ZOIA Pharma empowers patients on the journey towards better health and well-being, visit: zoiapharma.com.

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A Pentec™ Company

www.zoiahealthcare.com

MEET CLEMENTINE AND EVERETT

a story of hope in the face of a missed diagnosis

“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. Newborn screening tests for levels of methionine in the blood, which isn’t always elevated in the first 24-48 hours after birth. In addition, not all types of homocystinuria can be detected with elevated methionine.

Today, we want to share a story that illustrates advocacy, collaboration the profound impact of a missed diagnosis — Clementine and Everett’s Story.

HCU NETWORK AMERICA

Clementine was born into a family overjoyed with the birth of their first daughter, the sister of two devoted big brothers. After an uncomplicated pregnancy and birth, Clementine presented as a perfectly healthy and normal baby. When the pediatrician repeated the newborn screening blood spot at her two week check up the family knew the drill and didn’t think much of it.

Settling into a new routine, the family was shocked to receive the call stating her newborn screening was abnormal and they needed to bring her to Children’s Hospital Colorado right away.

They reacted as any parent would, with panic. They began googling what conditions were on the newborn screening and trying to piece together what could be wrong.

The following day, they took her for bloodwork, which revealed a homocysteine level of 153.9 mcmol/L and a methionine level of 736 nml/L. They did't know what that meant other than it was WAY above the normal range.

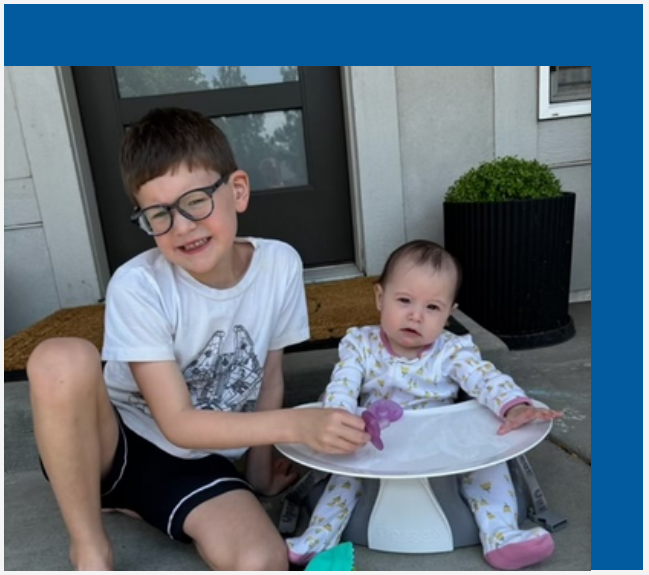
The family was frantic for more information. What did this mean? Was she in immediate danger? Was there treatment? A cure? What do we do? These are all the questions swirling in their minds.

Thankfully, their questions would be answered upon receiving a call from a genetic counselor at the metabolic clinic from Children's Hospital Colorado. She confirmed that Clementine was born with a rare metabolic condition called Homocystinuria. They were able to meet with the metabolic team the following day to discuss the diagnosis and treatment options.

The family learned that each of their other children would have a 25% chance of also having HCU. The family was confused because both their older boys passed newborn screening without issue. They thought they were in the clear. Both siblings had their blood drawn, and it was discovered that one of the other brothers also had homocystinuria with an astounding level of 222.7 mcmol/L.

The family was shocked, devastated, and distraught. How could this have happened? Everett missed the chance to be treated from birth. The family learned that the Colorado State Newborn screening labs' previous threshold for methionine was 100 mcmol/L.

> **Clementine and Everetts story is a testament to the importance of revised newborn screening for HCU patients and the collaboration that takes place between organizations, clinics and labs.**



After attending an event and learning about the issues related to HCU from HCU Network America's President, Margie McGlynn, the Colorado lab director, in collaboration with the metabolic clinic at Children's Colorado, lowered the threshold to 33! Everett was born when the threshold was higher and therefore missed on the screening. Clementine was the first baby born in Colorado to be diagnosed with a lowered threshold.

We are excited to announce that HCU Network America will be participating in the APHL Newborn Screening Symposium in Providence, RI. Our primary objective at this event is to bring awareness to state labs, the importance of lowering the methionine threshold, and educate physicians on recognizing the signs and symptoms of homocystinuria, as well as to provide guidance on patient referrals to genetic experts.

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 Clementine and Everett's. Together, we can make a meaningful difference in the lives of those affected by homocystinuria.

Matching Gift Challenge

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

During the winter holidays, warmth 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 Everett and Clementine's. Please share our appeal letter with your colleagues, friends, and family.

Three Easy Ways to Donate

Click here to donate

https://bit.ly/hcumatch25

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

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?

RACE RECAP

Team	Miles	Funds
Amy's Army	0	$2,973
Miles for Marley	31.5	$2,850
Miles for Andy	36.5	$561
Grayson's Gang	338.01	$780
Matteo's Muggles	137	$2,210
Classical Champions	472.41	$1,654
Miles for Marcus	34.44	$2,948
Renna's Rare Runners	60	$570
Team Recordati	664.17	$510
Carson's MTHFR Gene Team	0	$1,190
CanPKU+ Racers	140.66	$1,149
Team Lewis	92.6	$140

Team	Miles	Funds
Team Butter Stick	22.72	$5,525
Team Anniston	40	$1,530
Team Hunt for Research	245	$6,090
Team Will for HCU	139.49	$1,370
Brooke's Blazers	716.05	$1,840
Ellie's Entourage	59.39	$4,711
Team Hawkins	78	$300
Cure for Casey	2	195
Leo Frank	0	$20
Recordati-Sylvia's Supporters	1,467.99	$130
ASA Walkers	59.5	$260
March For Mariella	0	$460

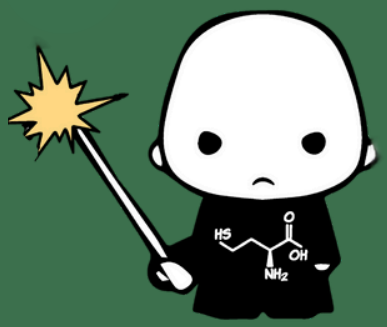
THIS SEPTEMBER

HCU HEROES

Race for Research

CURE

WALK • RUN • RIDE



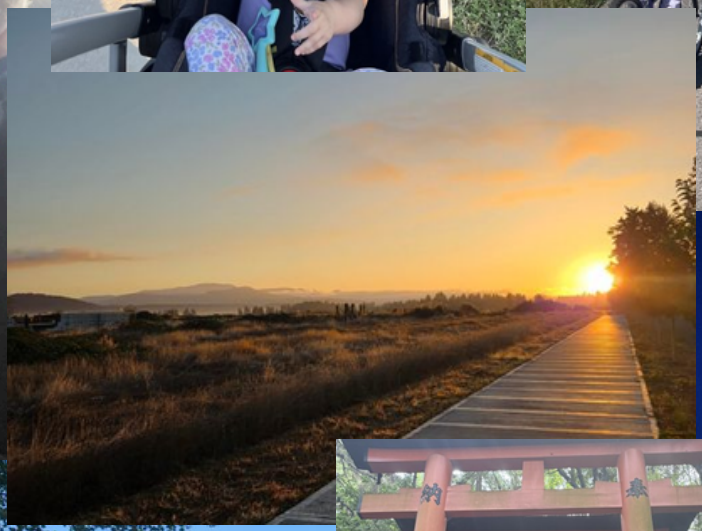
Thank You To Our Race For Research Sponsor!

RECORDATI RARE DISEASES

Focused on the Few®

HCU NETWORK AMERICA

THE WIZARDING WALL OF RACERS



Meet Our New Board Member!

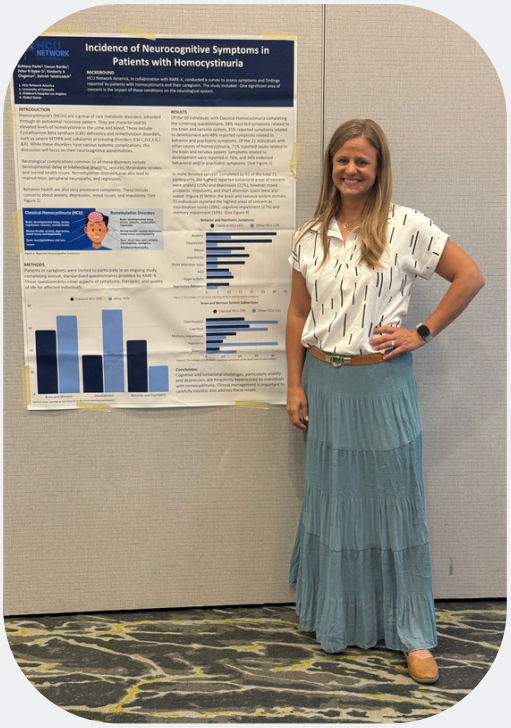
SCAN ME

Dr. Sagar Vaidya

SEPTEMBER RECAP

HCU Network America Was At...

Canpku Conference



Events like this are an incredible opportunity to raise awareness, share research, and collaborate with partners in the rare disease community.

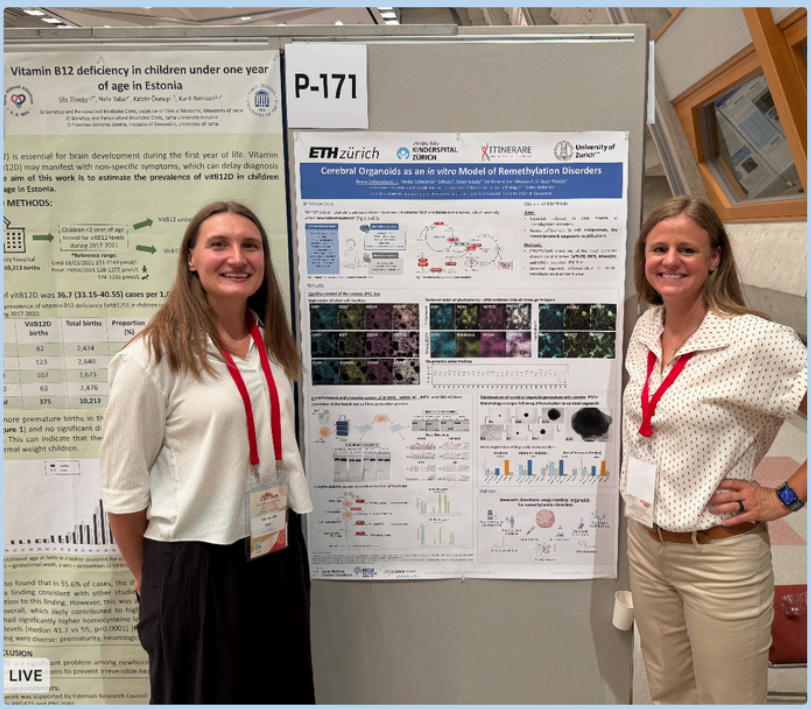
AMINO ALLIES
WWW.CANPKU.ORG

SEPTEMBER RECAP

HCUNA AT INTERNATIONAL CONGRESS OF INBORN ERRORS OF METABOLISM

KYOTO, JAPAN

We are so proud of Brittany attending AND presenting at ICEM. This was vital as we shared patient perspectives and connected with the global metabolic community. Together, we're accelerating progress for homocystinuria.



SEPTEMBER RECAP

North American Metabolic Academy

created by the Society for Inherited Metabolic Disorders

Danaé and Grace were able to chat with trainees in medical & biochemical genetics & share the resources and support that HCU Network America provides.

SIMD NAMA

Society for Inherited Metabolic Disorders
North American Metabolic Academy

Community in ACTION!

While in GA for NAMA, we had the opportunity to meet up with Ben & Lindsey from HC&U: A Homocystinuria Podcast as well as our September 2024 #HCUHero, Carter, and his amazing parents!
We love being able to catch up with the community when we are out on the road!

Georgia

SEPTEMBER RECAP

flok family Camp

East Coast. 2025

We are so grateful to have attended Flok Family Camp again! With a goal of getting patients and families together for not only educational sessions but some good family fun

It's A FAMILY AFFAIR

In The City of Brotherly Love

July 10-12, 2026
Philadelphia, PA

- Patient Focused Panels
- Breakout Sessions
- KidsZone and Teen Zone
- Community Building Opportunities

- Dedicated Science Day for Families and Professionals
- Scientific Poster Session
- HCU Hero Award Banquet
- Celebration Banquet in Honor of Margie McGlynn
- Networking Opportunities

Patient & Family Conference

REGISTER NOW

HCU NETWORK AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR

PUBLICATION FUNDED BY HCUNA

Givinostat rescues folding of cystathionine beta-synthase and ameliorates murine homocystinuria

ELSEVIER

Biochemical Pharmacology
Volume 239, September 2025, 117079

Givinostat rescues folding of cystathionine beta-synthase and ameliorates murine homocystinuria

Maria Petrosino, Karim Zuhra, Ela Mijatovic, Thilo Magnus Philipp, Olivier Bremer, Kelly Ascenção, Csaba Szabo, Tomas Majtan

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IN CASE YOU MISSED IT...

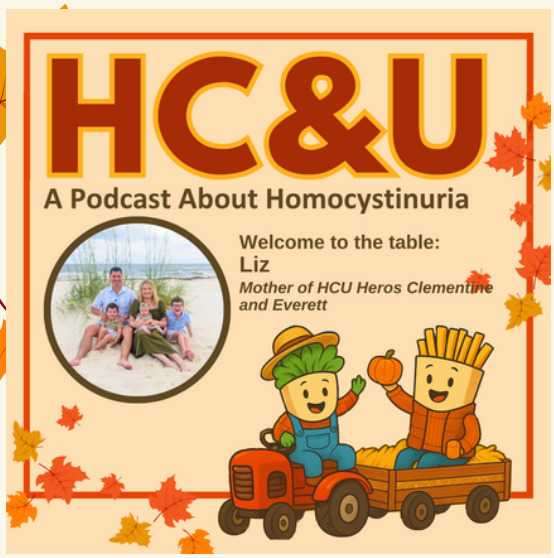
The HC&U Podcast is back!!!

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!

The latest episode



Ben welcomes Liz, mom of HCU Heroes Clementine & Everett.

Liz shares her family's powerful story — how newborn screening caught Clementine's HCU diagnosis, which ultimately led to Everett's own diagnosis. She opens up about the challenges, triumphs, and hope that fuel her advocacy for better awareness, research, and treatments for the HCU community.

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

LISTEN ON Spotify

Listen on Apple Podcasts

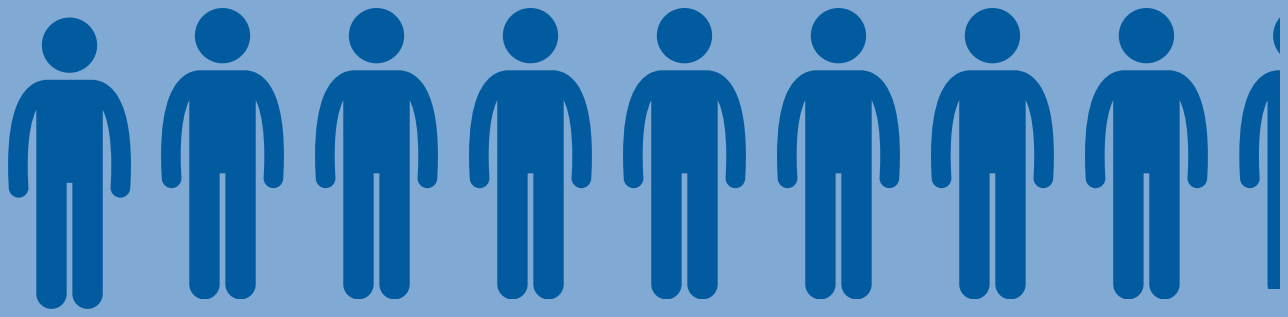
LISTEN ON iHeartRADIO

Listen on amazon music

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RARE X

Of the 81 participants who took the Head-to-Toe Survey
Here are the top 5 areas the data is pointing too



53
Vision Issues

48
Brain/Nervous System

36
Bones/Cartilage /Connective Tissue

36
Sleep

40
Digestive Issues

Complete the Head to Toe Survey at https://homocystinuria.rare-x.org

CANPKU+ CANPCU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

NEW COURSE AVAILABLE!

From Records to Research: Making Sense of Health Data for Rare Diseases

RDCA-DAP® | CRITICAL PATH INSTITUTE | NORD National Organization for Rare Disorders

New Resource

From Records to Research: Making Sense of Health Data for Rare Diseases.

Click Here

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*!

Contact Register

What?

- **What** is it?
  - A secure private survey for individuals or families affected by Homocystinuria
- **What** will I share?
  - Patient's birthdate, gender, exact diagnosis, and how they were diagnosed
- **What** will my info be used for?
  - Confidential and will not be shared unless we have permission
  - Helps HCUNA achieve our goals

Why?

- **Why** should I join?
  - Able to find other families and patients in your state and request contact information
  - Access to exclusive materials (ex: we may have a webinar that a presenter doesn't want to share publicly but is okay sharing with just our community)

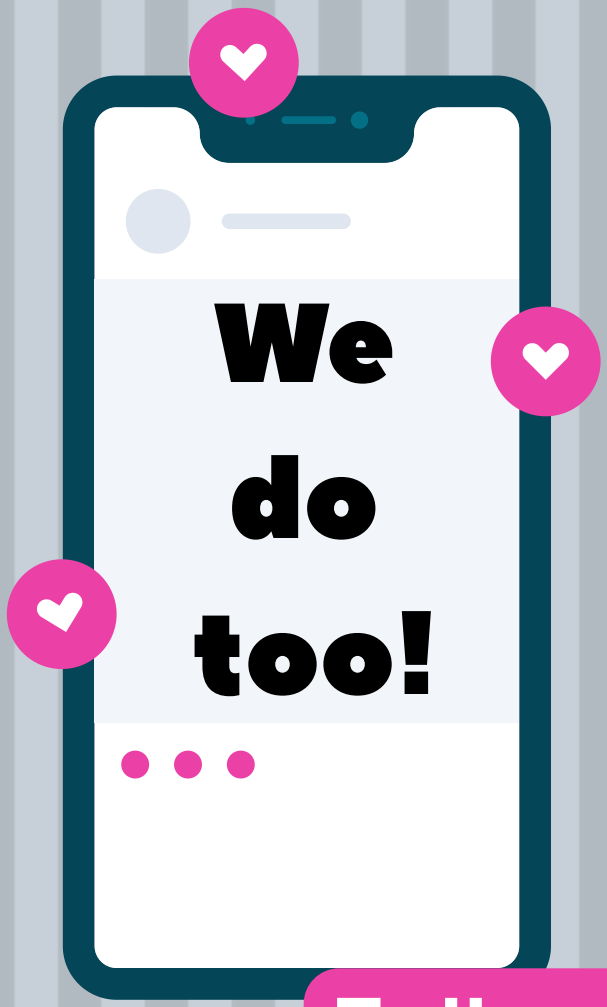
How?

- **How** do I participate?
  - The form takes 3-5 minutes to complete
  - Visit our website and click on "contact register" tab or...

Click Here

Follow Us!

Do you have social media??



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