The HCU Herald

Featuring...

HCU Hero
Casey from Illinois

February 2024

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

HCU HERO: CASEY FROM ILLINOIS

In September of 2022, the Shapiro family grew by one when we found out we were pregnant with our son, Casey. Our seven-year-old daughter, Lillian, was so excited to find out she had a baby brother on the way. Our family did all we could to prepare for our new baby, including genetic testing. We decided to see a geneticist because Casey's maternal grandmother and mom have severe anemia, to the point where mom had 11 iron infusions throughout pregnancy. All of our test results came back "normal", so we thought we had nothing to fear.

We welcomed Casey into the world on a beautiful, hot spring day in May 2023. Our delivery was smooth, and he latched instantly! Casey's newborn screening came back with flying colors, other than some minor jaundice at the time. The same thing happened with our daughter, so we were not alarmed. We were discharged from the hospital after only one night.

The summer kicked off with an active, healthy, breastfed baby that loved to smile, coo and be on his tummy. We played every day, traveled, camped under the stars and went to concerts. Our boy was hitting milestones and doing something new every day. As a stay-at-home mom and a dad who works from home, we relished watching Casey grow.

HCU HERO: CASEY FROM ILLINOIS

In August we started noticing signs of regression. Our three-month-old baby wasn't moving during tummy time, stopped tracking faces, hands or toys. By September he was fighting to stay awake, and his muscle tone had diminished. The jaundice had increased, and Casey was yellow all over. Our family doctor seemed positive it was a response to an ear infection. Little did we know, this was the start of Casey's illness.

When there was no sign of progress, we called our local hospital. Immediately the nurse said, “Drop everything and call 911.” My husband was calm but felt the urgency like the operator did, whereas I was still thinking this was due to the ear infection. Next thing we knew, we were in the back of an ambulance heading to Ann & Robert H. Lurie Children’s Hospital of Chicago. I kept thinking, *he has an ear infection, he's feeling the effects of medication, we will be right back home.* After two blood transfusions and a diagnosis of genetic anemia (that would later be discovered to be due to his Cobalamin), we came back home after a week-long stay in the hospital. At this point we thought the worst was over. However, four days later Casey began to decline again. He was losing weight and his family doctor deemed him “failure to thrive”.

In the month of October, we spent 20 consecutive days living on that gray couch in the hospital. It was during this stay where we experienced more trauma than words can describe. We learned Casey had a brain bleed that had started in September. He was placed on an NG tube, and it wasn’t until after every blood test, body scan, ultrasound, and X-ray was performed that we received a confirmed diagnosis of Cobalamin C Disorder (CblC).

HCU HERO: CASEY FROM ILLINOIS

We could barely process what any of this meant. We had more questions than we had answers. While CblC was explained to us, I kept hearing my husband's voice saying "We are built for this", while at the same time hearing the team of doctors saying, "this can cause seizures, blindness, heart attack and strokes". We could barely wrap ourheads around what we were up against. We will never forget looking at Casey laying in wires from his EKG, pulseOX, and NG tube and seeing him smile at us. The doctors said, "Casey is so lucky to have you both as parents." Suddenly our world shifted, and we prepared to fight alongside Casey and his lifelong disease.

The first concern from his care team was his beautiful blue eyes. CblC can cause Macular Degeneration and loss of vision. Thankfully, as of now, his eyes are in great shape. His brain and heart were next. Doctors determined his brain bleed was caused by pressure on his nerves, and luckily, he didn't need brain surgery. As for his heart, his EKG came back within the normal range as well. As testing continued and results came back positively, we still had millions of questions. We were still in shock, fighting sleep deprivation, breast pumping every two hours, and missing our daughter back home.

"I kept hearing my husband's voice saying "We are built for this.", while at the same time hearing the team of doctors saying "This can cause seizures, blindness, heart attack and strokes." We could barely wrap our heads around what we were up against."

HCU HERO: CASEY FROM ILLINOIS

As our stay in the hospital continued, my husband and I learned how to drop an NG tube. We watched cameras go down into our son's body and we learned to give injections from Casey's care team. We also learned how to grind medication and use IV poles to administer breast milk through this tube, all while still functioning on no sleep and continuing to be the best versions of ourselves. Looking back at it now, the time went by quickly, and we are so thankful for that. However, when we reflect on how we felt in the moment, the days felt like years.

When we returned home, all our family and friends rallied around us. They showed support with food deliveries, gift cards, care packages, visits, and lots of hugs and words of encouragement. Casey was still on his NG tube, and we started nightly injections, daily medications, and physical therapy once a week. We made a chart to keep track of medications and the phone didn't stop ringing for appointments. The one we looked forward to the most was Casey's final swallow study so he could come off the NG tube. After three weeks at home, Casey was off the NG and after two months at home, he was coming back to baseline and his care team was shrinking. We started to see a new version of him. He started to hit his milestones again. He was rolling, picking up his head, reaching for toys, laughing, and becoming more playful each day! We had finally settled into our new routine and began to feel confident in our abilities to tackle CblC.

HCU HERO: CASEY FROM ILLINOIS

However, another battle was around the corner once cold and flu season hit. CblC symptoms can present when the immune system is compromised. In December Casey contracted a common cold and began to have seizures. We were back in the hospital and Casey had his first EEG. This time, we worked as a well-oiled machine to aid Casey with his new diagnosis of epilepsy, which is another potential symptom caused by CblC. After four more EEGs we have stabilized the seizures with medication.

Casey is now eight months old. He is getting stronger every day, and as parents, we are trying not to overanalyze every move he makes. With a rare diagnosis such as this, it can feel like you constantly live in a place of fight or flight. We are flooded with worries like: “When is the next hospital stay?”; “Is he having another seizure?”; and “Are we doing enough?.

Thankfully, we have connected with other families in our rare disease community. We have found a lot of peace in reading other Hero Stories. We are always learning about Cobalamin Disorder and have immense gratitude for Casey’s entire care team. We love to see the excitement and joy in doctors' faces when they see Casey for a follow-up and he's gained weight, continues to do new things, and has hit new milestones! We are so proud!

HCU HERO: CASEY FROM ILLINOIS

My husband and I continue to care for Casey at home. He still receives daily shots and oral medications that are projected to continue for the rest of his life, or until research tells us otherwise. It's hard to predict where Casey will be in five years, but we are hopeful that he will one day attend public school with minimal impacts from CblC. Our biggest concerns are navigating him not getting sick and preventing what we call “CblC flare-ups”.

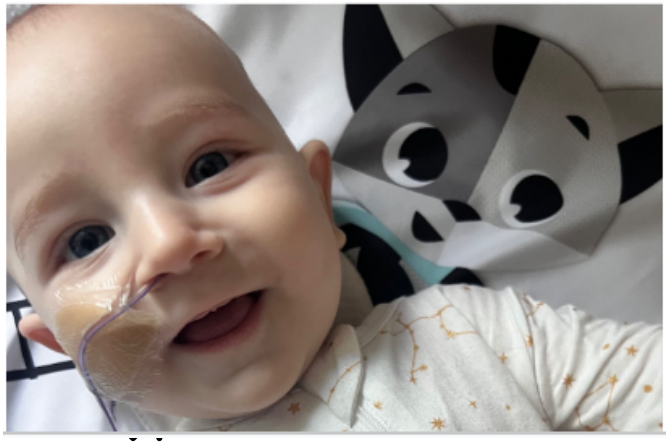
“It is important to continue to spread education, to keep the curiosity momentum going, and to encourage science to find a cure.”

Currently, Casey will sleep through his injections, and we are thankful for that. We have learned creative ways to give injections from other families with older children. However, we can never think too far into the future as we don’t know where his CblC journey will take him.

Casey Marshall Shapiro is the bravest boy we know. He is loved, supported, and now all we can do is be HOPEFUL that we find a cure. We appreciate our family and friends who have taken the time to learn about his disease and check in with us. A lot of our family members have embraced the zebra ribbon to support Casey's rare disease diagnosis. It is important to continue to spread education, to keep the curiosity momentum going, and to encourage science to find a cure. We are thankful for the resources from Casey's care team who have mentally and emotionally cared for our family through this battle.

We were scared to share Casey's story because we felt so alone. Slowly, we started to connect with other families, members of our community who offered resources, and friends who were willing to drop off a crock pot of our favorite meals. We started to feel less alone, and more seen.

If we had to give advice to a new member of the HCU/CblC family, we would say to give yourself *grace*. This diagnosis can be shocking, but you're not alone. Take each day one step at a time, speak with your care teams, seek therapy, connect with other families, and SPEAK OUT on social media. Turn inward to your family, don't be afraid to cry, or send questions to your doctor on MyChart at 3 am. Remind yourself that while this is life-changing and you may need to adjust your plans, you can still go out and see the world and enjoy life. Even when in survival mode, look for the good. No amount of answers will take away the feelings you may have to want to control this. ACCEPTANCE IS KEY! For injections, find the band-aids that make you laugh. Call your EEG a "fun ponytail", search for the best vending machines in the hospitals, and learn to laugh again. Your family will get through this. There is a community out there, and if you're reading this, *you've found it*.



To read Casey's story on our website, visit:
https://hcunetworkamerica.org/casey-illinois/

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

Meet our Medical Advisor

MEET OUR MEDICAL ADVISOR

Curtis R. Coughlin II, PhD, MS, MBE

Curtis Coughlin is an associate professor at the University of Colorado Anschutz Medical Campus. He was trained as a genetic counselor at the Beaver College Genetic Counseling Program and worked in inherited metabolic disease clinics both in Philadelphia and Colorado. He completed his doctoral training with Professor Stephen Goodman at the University of Colorado, and his basic science research has continued to focus on inborn errors of metabolism. The overall aim of the Coughlin laboratory is to leverage our understanding of biochemistry to improve the diagnosis and treatment of these genetic disorders. Dr. Coughlin has led several internal studies focused on the natural history and impact of treatment on inherited metabolic disorders such as pyridoxine-dependent epilepsy and glutaric aciduria type I. He is committed to partnering with families, clinicians, and scientists with the primary goal of ensuring scientific advances are relevant and patient centered.

Meet our Treasurers

MEET OUR NEW TREASURERS

Darren,
Treasurer

Dana,
Assistant Treasurer

Darren and Dana Hunt reside in Overland Park, Kansas with their sons Ethan and Carson. They are thrilled to continue their work with HCU Network America, as members of the board, and now, treasurers. They are passionate about spreading awareness and raising funds for our community!

Update: Newborn Screening

UMass Chan MEDICAL SCHOOL

New England Newborn Screening Program
University of Massachusetts Chan Medical School
377 Plantation St. Biotech 4: 2nd floor
Worcester, MA 01605
774-455-4600 (office)

Project Summary

Devinder Kaur, Ph.D, Lead Scientist, UMass Chan MEdical School

Development of Reference Ranges for Additional Newborn Screening Markers for Early Detection of the Homocystinurias: Classical Homocystinuria and Remethylation Disorders

PI: Devinder Kaur, Ph.D., Lead Scientist, UMass Chan Medical School, Worcester, MA

Background: Newborn screening (NBS) for the homocystinurias (HCU), a group of disorders, has traditionally relied upon abnormal levels of the primary markers methionine and propionylcarnitine (C3) in dried blood spots (DBS) of newborns. These markers are not specific for these conditions because the abnormal result can also be due to secondary dietary interventions, and other diseases, thus causing false-positive results. This leads to unnecessary follow-up of unaffected babies, parental anxiety, and unneeded medical tests. The NBS programs attempt to minimize the number of false-positive test results by using more specific markers and/or second tier confirmatory testing.

More specific markers for HCU are total homocysteine (tHcy), methylmalonic acid (MMA), and methylcitric acid (MCA). Analysis of additional markers such as cystathionine (cysta) and cysteine (cys), along with tHcy, MMA and MCA in the DBS may further improve the differentiation amongst the various forms of HCU. However, NBS for HCU using these markers as primary and/or 2nd tier test (2TT) is still not widely established due to the cost of instrumentation, complexity of test, limited capacity, and resources. We embarked on a pilot project to develop and validate a simple, robust, and high throughput liquid chromatography tandem mass spectrometry (LC-MS/MS) 2TT test in particular, measurement of tHcy, cystathionine, cysteine and MMA.

The next step is to determine a reference range and cutoff value for the target markers. A reference range for a screening test is a range of values that is considered normal for a particular test in a healthy population. The reference range helps healthcare professionals determine whether an individual's test results fall within expected values, and if a newborn is at risk for a disorder. NBS programs, including New England Newborn Screening (NENSP), have implemented different cutoffs for some metabolic disorders detectable by tandem mass spectrometry (MS/MS) depending on the infant's age (in hours) at blood collection. In addition, co-variates such as prematurity, birth weight, transfusions, neonatal jaundice, type of feed and parenteral nutrition can all potentially influence NBS results and need to be considered when establishing cut-off values and interpreting results.

Update: Newborn Screening

UMass Chan MEDICAL SCHOOL

New England Newborn Screening Program
University of Massachusetts Chan Medical School
377 Plantation St. Biotech 4: 2nd floor
Worcester, MA 01605
774-455-4600 (office)

Goals and Objectives: The overall goal of this project is to develop, optimize and implement new 2TT algorithms in order to assess the applicability of this approach in lowering the number of false positives while increasing the likelihood of identifying newborns with classical homocystinuria (HCU) and HCU-Remethylation disorders. We received funding from HCU Network America to accomplish the following objectives:

- Determine normal reference ranges for total homocysteine (tHcy), cystathionine (cysta), total cysteine (tCys) and methylmalonic (MMA) by analyzing >200 DBSs collected between 24-48 hours of age, from babies thought to be negative for these conditions, using the newly developed method.
- Retrospectively analyze confirmed classical HCU and HCU-Remethylation (Remet) disorders DBS.
- Analyze data, determine reference ranges, and publish findings.

Results: We have developed and validated a high throughput 2TT LC-MS/MS assay to simultaneously measure elevated tCys, tHcy, MMA and Cysta using a single DBS for screening HCU disorders. This method was used to determine reference ranges of >400 DBSs collected between 24-48 hours of age from babies thought to be negative for these conditions. The preliminary cutoffs were determined at the 99th percentile for tCys, tHcy, MMA and Cysta by comparing the control group with confirmed classical HCU and HCU-ReMet disorders DBS. This assay clearly differentiated biochemical patterns between confirmed cases of classical homocystinuria, methylmalonic acidemia, Methylene tetrahydrofolate reductase deficiency, cobalamin C deficiency and propionic acidemia.

Future Plans: The analyses performed by NENSP thus far have been on retrieved specimens. We will re-evaluate and adjust the reference ranges and cutoffs when the method is implemented and the sample size increases. We anticipate that implementation of 2TT will also help us assess and refine our current cutoffs for primary markers and the metabolic profile. Further, the two-tier strategy will offer the advantage of reducing the burden of both false positives and false negative results.

Thank you for supporting our Double Good Popcorn Fundraiser!

FUNDRAISER SUMMARY

TOTAL SALES
$1,336.00 ($668 to HCUNA)

STORES WITH SALES
4

STATES REACHED
10

SUPPORTERS
25

HCU NETWORK AMERICA

Raising Joy Double Good

BAGS DONATED
8

DONATION IMPACT
With over 5 million bags donated, each kernel has brought joy to educators and healthcare heroes in all of our communities.

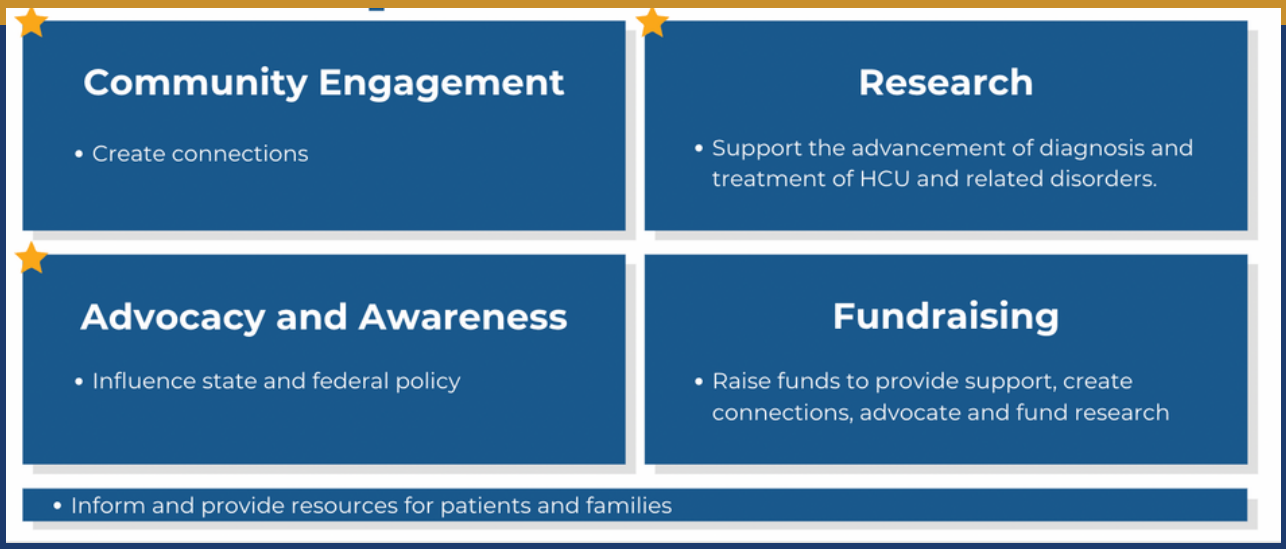
The funds raised will benefit our 2024 Family Conference in Aurora, CO!

Event Recap / Join our committees!

Missed our Community Town Hall Meeting?

Catch the replay here!

We'd love for YOU to join one of our newly formed committees!



Fill in the Committee interest survey here:
https://www.surveymonkey.com/r/HCUNAVolunteer

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 catch it on Spotify or Apple Podcasts!

LISTEN ON Spotify

Listen on Apple Podcasts

The latest episode

The episode features Brittany Parke, mom of Grayson, who lives with Homocystinuria, Cobalamin G. Brittany gives insight into why advocacy & research for Hydroxocobalamin is important to the HCU cobalamin community!

Registration:

TICKETSIGNUP.IO/TICKETEVENT/HCUCONFERENCE

MOVING MOUNTAINS

Classical HCU | Cobalamin Disorders | Severe MTHFR

Patient-Family Conference

Aurora, Colorado

June 29-30, 2024

HCU NETWORK AMERICA

- KEYNOTE FOCUSED ON MENTAL HEALTH
- AGING WITH THE HOMOCYSTINURIAS PANEL
- BREAKOUT SESSIONS
- DIAGNOSTIC TOOLS AND NEWBORN SCREENING
- RESEARCH UPDATES
- HCU HERO AWARD RECEPTION

Scholarship opportunity!

flok The Guthrie-Koch Scholarship

- **Do you have Classical HCU?**
- **Are you a *high-school senior* or *current student* pursuing an undergraduate degree or technical school?**

If you answered 'yes', you are eligible to apply for the Guthrie-Koch Scholarship Program!

The Guthrie-Koch Scholarship Program was founded in 1997 to recognize outstanding young adults with PKU pursuing higher education and provide financial support to these efforts, but has now been expanded to include young adults with Classical HCU and other metabolic disorders!

Applications for the 2024-2025 academic year are now being accepted at scholars.flok.org.

The application deadline is March 15, 2024.

GET YOUR SQUARES NOW!!!!

FOOTBALL SQUARES FUNDRAISER

FEBRUARY 11 | 6:25 PM ET

$10 PER SQUARE

4 CHANCES TO WIN!

VENMO: @TOM-HAWKINS-1

EMAIL TOM AFTER TO RECEIVE ACCESS TO BOARD!
TMMYHWK09@GMAIL.COM

$100 PAYOUTS 1ST-3RD QUARTER
$200 PAYOUT AT END OF THE GAME

OUR SUPER BOWL SQUARES FUNDRAISER WILL HELP TO FUND OUR 2024 FAMILY CONFERENCE IN AURORA, CO!

PANDA EXPRESS CHINESE KITCHEN®

+

HCU NETWORK AMERICA

When?

Friday, February 9, 2024

Place your online order for pickup or delivery on Friday, February 9!

Where?

Panda Express locations nationwide

www.pandaexpress.com

How?

Online orders only.

Apply code **920234** in the fundraiser code box during online checkout at www.pandaexpress.com or via App

28% of sales will be donated to HCU Network America!

Advocacy opportunity!

Apply for Travel & Lodging Assistance!

- Plane ticket on a commercial airline up to $450/person
- Mileage Reimbursement up to $200
- Complimentary flight through our travel partner Angel Flight West (restrictions apply and additional information required through application process)
- Up to 2 nights stay at the Conference Hotel
- Bus or Train Fare

Application deadline is February 6, 2024 at Midnight ET!

This event will bring together the incredible rare disease community for a day filled with learning, networking, and relationship-building.

The Living Rare Forum is an opportunity for people living with rare diseases and their families to come together, often for the first time, to gain practical knowledge on how to manage their health and live their best rare lives.

Program topics often include sessions about how to advocate for yourself in the healthcare system, how to coordinate your medical care team, and how get involved in rare disease research and advocacy.

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**

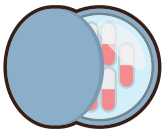
**Kits can only be sent to patients in the continental US. However, we are happy to connect virtually and share the educational materials with you via weblinks*!

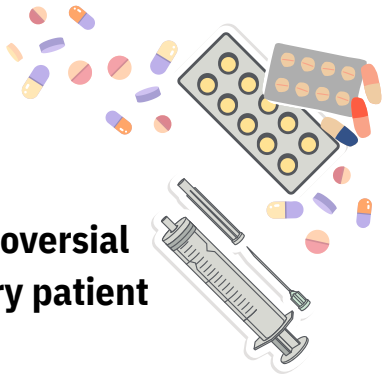
HOMOCYSTINURIAS

DATA COLLECTION PROGRAM

Medication and Supplement Usage Survey

Take the survey now at: homocystinuria.rare-x.org





Each type of Homocystinuria has published Guidelines for Diagnosis and Management (see below). Some therapies are more controversial and even with those that are accepted, every patient responds differently.

Take the Medication and Supplement Usage Survey and let us know about the Medications and Supplements you are prescribed - both for HCU and otherwise!

	Drugs with proven clinical benefit	Treatments without proven clinical benefit	To be avoided
Classical HCU	B6 (responsive patients) Betaine Medical formula Low protein diet B12 (only if deficient) Folate (only if deficient)	Cysteine	Fasting Nitrous oxide
Cobalamin related remethylation disorders	OHCbl parenteral Betaine	L-Carnitine Methionine	Nitrous oxide Protein restriction
MTHFR deficiency	OHCbl parenteral Betaine	Folinic acid/ 5-Methylfolate* L-Carnitine Methionine*	Nitrous oxide Folic acid Protein restriction

***Has been applied with clinical benefit in single cases**

Morris, Andrew A M et al. "Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency." Journal of inherited metabolic disease vol. 40,1 (2017): 49-74. doi:10.1007/s10545-016-9979-0

Huemer, Martina et al. "Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency." Journal of inherited metabolic disease vol. 40,1 (2017): 21-48. doi:10.1007/s10545-016-9991-4

CANPKU+ CANPCU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

RARE DISEASE WEEK ON CAPITOL HILL

Registration is open!

Rare Disease Week on Capitol Hill empowers and inspires hundreds of advocates each year. The connections you make during the week will impact rare disease patients for generations to come.

Hosted by the Rare Disease Legislative Advocates (a program of the EveryLife Foundation for Rare Diseases), this multi-day event brings together rare disease advocates from across the country to make their voices heard by their Members of Congress. Participants are educated on policy proposals impacting the rare disease community and provided opportunities to advocate for policy changes directly to their Members of Congress. No matter one's connection to rare disease or their advocacy experience level, all are welcome.

Click here to learn more, and to register to attend!

LET US KNOW!

Planning to attend Rare Disease Day events?

We'd love to connect you with others in our community who will be attending!

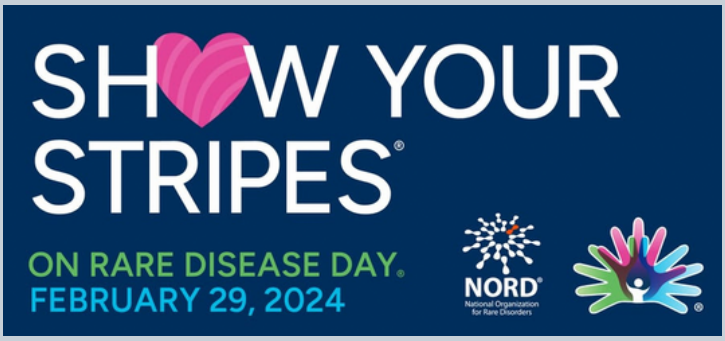
RARE DISEASE WEEK EVENTS

Rare Disease Day @ NIH

When? Thursday, Feb. 29, 2024, from 9 am-5 pm EST
Where? NIH main campus (Natcher Conference Center)
OR virtually

The event aims to raise awareness about rare diseases, the people they affect, and NIH collaborations that address scientific challenges and advance research for new treatments.

Click **here** for more info & to register!



NORD - #ShowyourStripes at the Today Show Plaza!

When? Thursday, February 29, 6:30 am EST
Where? The Today Show!
Rockefeller Plaza, 35 West 48th Street
(between 5th & 6th Avenues)

Get your posters ready, dress in your most festive zebra gear, and let's raise awareness for rare diseases. Share your pics with #ShowYourStripes and #RareDiseaseDay.

Click **here** for more info & to register!

FDA's Rare Disease Day 2024

When? Friday, March 1, 2024,
from 9 am-4:30 pm EST
Where? Virtually

Panel Discussion topics will include:

- The legal framework for approving studies and medical products at FDA
- Decentralized clinical trials and digital health technologies
- Where to find important information and documents related to clinical trials
- Legal and ethical requirements for consent forms in clinical trials
- FDA initiatives to advance medical product development for rare diseases
- Ways for patients to engage with FDA

Click **here** for more info & to register!

Rare Disease Day is coming...
Grab your Gear!

Grab your gear!

https://www.bonfire.com/store/hcu-haberdashery/

Order by February 5 to receive in time for Rare Disease Day!

Acappella study on Classical Homocystinuria

Now enrolling

Sponsor: Travere Therapeutics
Study duration: About 6.5 years
Study type: Natural History (no investigational medicine given)
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)	1 - 65 years of age	The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

**You (or your child) will need to meet all other study criteria to take part in the ACAPPELLA Study.*

Study locations

Colorado – Washington DC – Georgia – Indiana – Massachusetts – Pennsylvania
Ireland – United Kingdom

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

For new participants, we now have an option for the study to come to you! (decentralized site). Please inquire to learn more.**
**restrictions apply

You may be able to receive payment for time and travel when you participate in this study.

Talk to your doctor and family members about 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

Questions?
Email: medinfo@travere.com

For more information, please scan the QR code or visit:
www.hcuconnection.com

MA-PE-23-0069 Nov 2023

TRAVERE® THERAPEUTICS

UPDATE

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

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

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.

MEDICAL ASSISTANCE

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

Contact Register

What is the contact register?

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

What will this information be used for?

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

How do I participate?

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

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