The HCU Herald

Featuring...

HCU Hero
Nino from Minnesota

December 2023

HCU NETWORK AMERICA

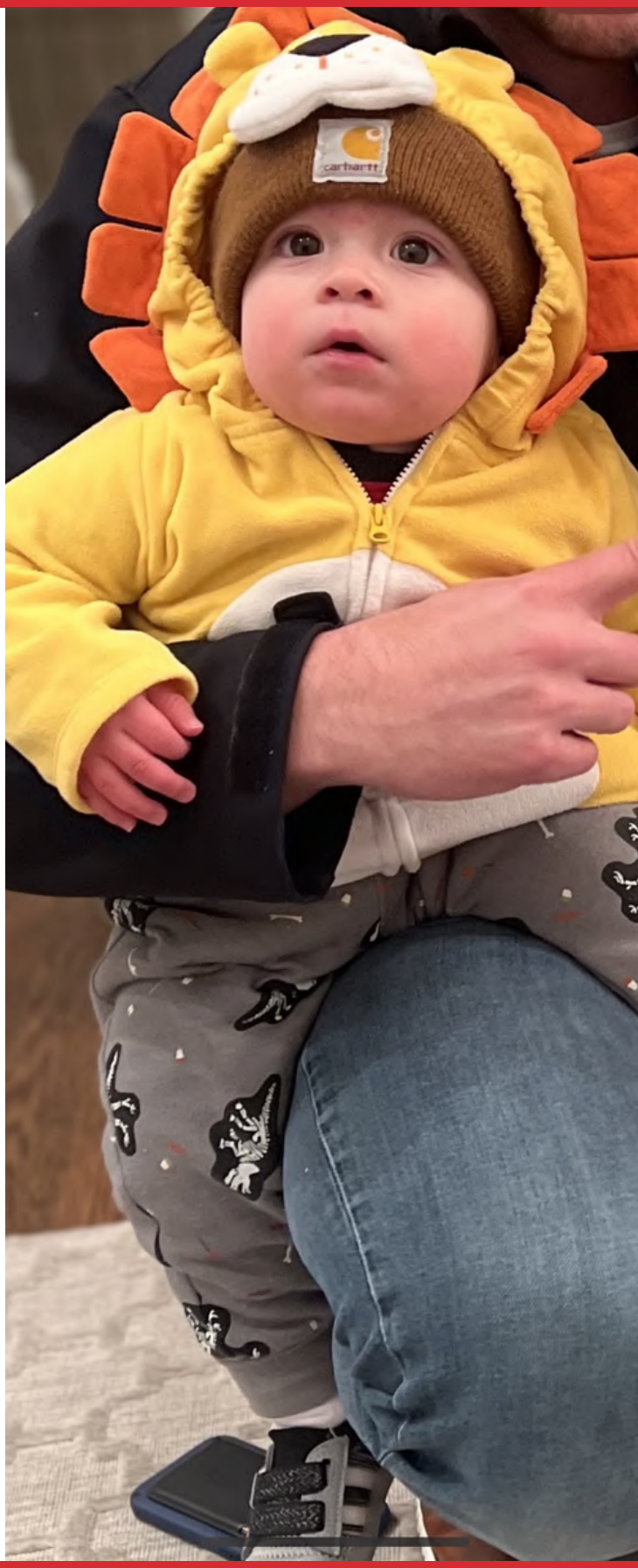
All things Homocystinuria: patient stories, resources, research, events and more!

HCU HERO: NINO FROM MINNESOTA

Our little Nino was the second baby to join our family. He was born in October 2022, following an uncomplicated pregnancy. We were excited to grow our family and give our first son a brother - we knew the two siblings would be two peas in a pod. And indeed, they were.

Nino's delivery felt completely textbook. He was born via scheduled c-section because mom had a complicated first delivery and it felt the safest route. So, Nino was brought into the world in a very quick and anticipated manner. From the moment he emerged, everything felt good and joyous, and we focused on recovery and getting home.

Like all newborns in Minnesota, Nino was privileged to partake in newborn screening. We didn't know at the time that it was an absolute blessing - in fact, mom hated to see that first blood draw, not knowing that there would be many more, with other more difficult tests and visits to the hospital. Nino did not express any symptoms of having any genetic disease, but soon enough, a geneticist reached out. In fact, the communication was slightly more shocking, as the pediatrician called dad and the geneticist called mom.



HCU HERO: NINO FROM MINNESOTA

We held Nino tight and with tears in our eyes made our way to the Minnesota Children's Hospital soon thereafter for a more comprehensive introduction to the wide realm of possibilities (don't worry, this story will have a happy ending!). The staff generally suggested that Nino might be suffering from one of the homocystinurias, and although they didn't have clarity on which one, began to administer treatment immediately. After some additional testing to determine the biomarkers were indeed persistent, we pushed on for genetic testing.

The entire family leaped at the chance to help narrow down Nino's diagnosis. We fleshed out the family tree, calling on relatives to narrow down information and family myths to provide a good quality history, and then all Nino's grandparents and parents jumped at the chance to send in DNA. With the influx of support, Nino was officially diagnosed with Cobalamin C deficiency.

Beginning treatment was difficult, but Nino deserved for us to remain committed to providing him with consistent medications and stay on top of doctor's appointments. His eyes and his heart were top priorities, and ensuring their best care became part of our routine. The blessing of newborn screening and immediate medical intervention is that Nino was able to receive the benefits of treatment right away, and we remain watchful of our happy, spirited, 13-month-old.

While we had the benefit of timeliness, Nino's initial diagnosis did not come with any certainties. We knew the absolute best-case and worst-case scenarios and maddened ourselves with the "what ifs?". Now that he was in our arms, the worst-case scenario was unimaginable. Case studies, trials and research papers became our life, and we spent months reviewing everything we could get our hands on. Still, it wasn't enough.

Our motivation to find clarity for Nino on his potential future brought us to the Mayo Clinic. We transferred his records and found comfort in the genetics team there. Nino was in GREAT hands. We were able to get some good answers about the exact type of Cobalamin C deficiency and what to expect - so far, the doctors have been spot on.

We are raising Nino in a house full of love and laughter. We know that he'll have a childhood filled with friends, family and joy. There will be ups and downs that we cannot foresee, but we will not stop advocating for our little guy or others like him. We cannot imagine where we would be without newborn screening and the critical advantage it brings families around the world. The benefits of beginning treatment early cannot be understated, and we are blessed to see those benefits with our son today.

To read Nino's story on our website, visit https://hcunetworkamerica.org/cobalamin-patient-stories/nino/

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

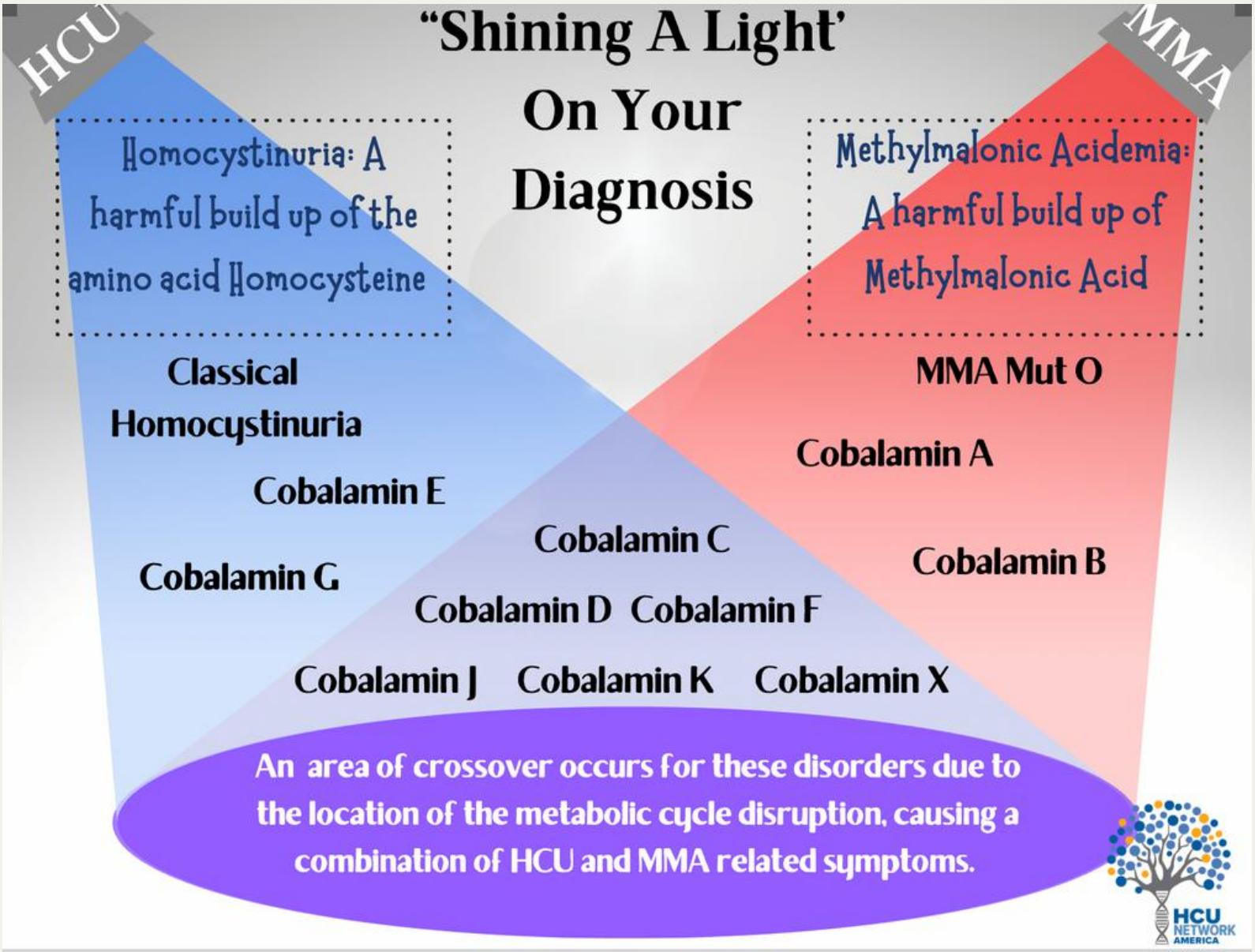
We are looking for new community members to join our fundraising team!

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

New resource



The metabolic pathways for the homocystinurias (HCU) and methylmalonic acidemias (MMA) are very confusing! While a majority of the cobalamin disorders are a combination of both HCU and MMA, not all are. This leads to a very distinct different type of treatment protocol for those who have a combined form. Knowing the type of Cobalamin disorder you have is important to ensuring your treatment best suites you!

Click here to download this new resource!

New resource

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*!

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!

IRB Approved - Study 00024964

OHSU MOLECULAR AND MEDICAL GENETICS

Has the national formula supply shortage impacted you or your family?

Laura Sliwoski is a Metabolic Dietitian with Oregon Health & Science University in Portland, Oregon. She is studying how the national formula shortage impacted patients with inborn errors of metabolism (IEM) and their families.

Your input can help identify measures to minimize or prevent the risk of future shortages.

You are invited to take part in an online survey where you will answer questions about your experience during the formula shortage. This includes questions about nutrition, stress levels because of the shortage, and the difficulty of switching to a different formula.

Who is eligible?

- Parents/caregivers of a child less than 18 years of age with an IEM
- Parent/caregiver of an adult with an IEM who is decisionally impaired

This survey will take about 15 minutes to complete. As a thank you for your time and participation, you have the option to be entered in a raffle to win a $100 Visa gift card.

To learn more about this study, scan the QR code below or visit https://redcap.link/6od39e6q. For questions, or to speak with the study team, please call 503-494-3137.

We appreciate your interest in our study!

OHSU

UPCOMING EVENTS

HOMOCYSTINURIA TEEN MEETUP

Our teen (10-22) meetups are an opportunity for teens with any of the homocystinurias to connect, learn, and share experiences.

DECEMBER 9, 2023
MEETUP AT 3 PM ET

MEET YOUR TEEN LEADER: LANDON

Landon is a 16-year-old with Classical HCU from West Virginia. He enjoys theater, his adorable puppy Milo, and getting to meet new people!

Register:
https://bit.ly/hcuteen

2024 Family Conference

Click here to register to attend!

Join us in June 2024! This oppourtinuity to learn, connect, and be supported is open to families of ALL of the homocystinurias! We hope you'll be there!

LO PRO HOLIDAY DESSERT

Chocolate Celebration Cake

Serves 12

ingredients

- 1-15.25 oz package chocolate cake mix
- 4 tablespoons instant chocolate pudding mix
- 12 ounces cola
- 3 ounces water
- 1-16 oz container frosting flavor of choice

NOTES:

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FOR CUPCAKES, FILL EACH TIN HALFWAY. RECIPE WILL MAKE 24 CUPCAKES. PROTEIN AND AMINO ACID VALUES WILL BE HALF.

~

SIZE OF CAKE WILL VARY BASED ON PAN USED. REMEMBER TO DIVIDE EVENLY INTO 12 SERVINGS.

directions

- Heat oven to 350ºF.
- Using the instructions on the cake mix box as a guide, select your baking pan, then grease and flour it.
- Combine cake mix, pudding mix, cola and water in a large bowl. Stir with a spoon or beat with a mixer for 2 minutes.
- Bake according to times on cake mix package based on your pan used. Cake is finished when a toothpick inserted comes out clean.
- Cool cake completely before frosting. Divide cake into 12 equal servings.

KCAL: 340 PROTEIN: 1 G ARGININE: 88 MG METHIONINE: 31
DETERMINED BY NUTRITIONIST PRO.

THANK YOU TO THOSE WHO PARTICIPATED IN OUR THANKSGIVING DAY FOOTBALL SQUARES FUNDRAISER!

WE RAISED $1,775 FOR OUR PROGRAM FUND!

THESE FUNDS WILL HELP TO MAKE OUR 2024 FAMILY CONFERENCE A REALITY!

Our winners were....

-Q1 : Chris Hummel

-Q2 & Q3: Danae Bartke

-Game winner: Ed Hempling

Join us for SUPER BOWL Squares on February 11, 2024!

OUR MATCHING GIFT IS BACK!!!

That's right, you heard us right! Thanks to three anonymous donors, **any funds** you help raise from **October through December 31, 2023**, will be matched **up to $25,000!**

We are asking every patient and family to help us raise funds for homocystinuria. During the winter holiday's warm hearts and generosity can be felt near and wide. During this time, we ask that you share our appeal letter with your colleagues, friends, and family.

See our appeal letter on the next two pages, or you can print it from here.

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?

15 S. Mallory Ave
Batavia, IL 60510

(630) 360-2087
info@hcunetworkamerica.org
http://www.hcunetworkamerica.org
Tax ID Number: 81-3646006

Dear Community,

As we near the end of 2023, I wanted to take a moment to express my deepest gratitude for your continued support and generosity throughout the year. Your contributions as a loyal HCU Network America donor directly impact the lives of people born with homocystinuria and enable the organization to help families navigate their condition and advocate for better practices.

HCU Network America is here to help this special group of people with the support and resources they need to navigate daily life. But we can't do it without your help! As a 501(C) (3), we need your donations, which are tax deductible, to continue with our mission and meet our goals. Your contribution can make a huge difference in the lives of all HCU patients and their families.

We would like to share an excerpt from one family's story:

> *When Amy was still a young child and before receiving a proper diagnosis, one neurologist suggested that she might be having "mini-seizures" when it appeared that she was staring into space. She had an abnormal EEG in the office, showing micro-seizures when provoked, but a 72-hour video EEG was normal. She continued to walk with severe "in-toeing" but the pediatric orthopedic thought she would grow out of it. Similarly, her pediatrician was unconcerned. In April of 2020, Amy had her first focal seizure while on a walk with her dad. She collapsed and was staring at the sky and was unresponsive to her dad's attempt to get her attention. After about 3-4 minutes, she came to, and he was able to support her enough to get her home. She was diagnosed with epilepsy and began taking medication to control the seizures. We also had her tested for autism, and she was diagnosed with that in 2021. A few months later she had another seizure during which only half of her body was seizing. There is some debate among her doctors as to whether this was actually a stroke. Around this time her unusual gate became more pronounced. In early 2022 we did genetic testing and learned about the Severe MTHFR deficiency. Her homocysteine level was over 200. It is better controlled now, with her numbers between 70-80. However, in May of 2023, Amy told us that she was having trouble seeing and that she was seeing "dots". After a visit to a retina specialist, we were told that her optic nerve was swollen and that we should take her to the ER. She ended up having a spinal tap to relieve the intracranial hypertension.*
>
> *It is hard to write about how having a child with special needs impacts the entire family, although the effects are considerable. We struggle to practice patience, with our children and ourselves, knowing that we are all experiencing a sense of loss. Our other children have had to be independent, and while ultimately this may benefit them in some ways, losing time with parents because they are busy with caretaking and the logistics that come with frequent therapies and doctors' appointments will no doubt have impacted them, as being independent and being forced to be independent are very different situations. We are so proud of Amy. She is our HCU hero because she demonstrates strength on a daily basis when she takes her medicine and suffers from its side effects without complaint. She has accepted her condition with grace and by example, teaches us to practice patience and inclusivity.*

HCU Network America was there to support this family at one of the most devastating times in their life. Since our inception in 2016 we have communicated with metabolic clinics all over the country to reach out to new patients and provide them toolkits which are filled with helpful tips and guidelines for living with HCU. We also financially support research that can help find new treatments!

Here are some of the highlights that your donations helped with over this past year:

- Created over a dozen resources to aid those living with HCU in their transition to adulthood.
- Expanded our NORD Medical Assistance Program to cover medical formula in addition to low protein foods.
- Hosted 3 round table discussions with newborn screening stakeholders across the country with over 80 participants. Here we shared problems in our newborn screening system and potential solutions.
- Published our first paper with data from the HCU Data Collection program, powered by Rare-X.
- Presented a poster on *Understanding the Burden of Classical Homocystinuria (HCU) from the Patient's Perspective: A Qualitative Study* at the annual symposium for the Society for the Study of Inborn Errors of Metabolism (SSIEM).
- Awarded our first grants for Cobalamin G and Severe MTHFR
- Hosted an Externally Lead, Patient Focused Drug Development (ELPFDD) meeting with key stakeholders, including the FDA.

Our goal this year is $50,000. Thanks to an anonymous donor, any funds you donate up to December 31, 2023 will be matched up to $25,000. In addition, if your employer matches charitable donations, they will match those too! If you personally know a patient with HCU, you can donate in their honor. We need your help, and appreciate **any** donation.

Take a minute to look at our website to see what we are up to and meet some of our "heroes": https://hcunetworkamerica.org. You can donate through our website or by mail.

Thank you once again for your unwavering support. Together, we can make a lasting impact and bring about positive change in the lives of those who need it most. Wishing you and your loved ones a joyful holiday season and a prosperous New Year.

With heartfelt gratitude,

Danae' Bartke

Danae' Bartke
HCU Network America, Executive Director

Donor levels:

- **Leadership Circle** - $5,000 or more
  Donor's name, HCU Patient's name and photo on homepage of website, along with certificate donation
- **HCU Champion** - $1,000 or more
  Donor's name, HCU Patient's name and photo on HCUNA donation page along with donation certificate
- **HCU Supporter** - $500 or more
  Donor's name, HCU patient's name on website, along with a donation certificate
- **HCU Ally's** - $100 or more
- Donor's name and HCU patient's name listed on website

4 ways to donate:

1. Use the enclosed slip and envelope
2. Go to https://bit.ly/donate-hcu
3. Text HCU2023 to 44-321
4. Scan the QR Code

iGive.com

You Shop. Your Charity Gets Money. For Free.

Your holiday shopping makes a difference

Shop at any of the 1,800 stores that want to help, and **HCU Network America** will receive a portion of your purchase! (On average 3% - (AmazonSmile was only half a percent!). Stores pay for it all. Never pay more, and sometimes less with coupons and deals.

Want to make your purchases count?

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

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

Shop normally (no special codes, no special anything) at any of about 1,800 stores. The Button is working in the background to let them know you're helping when you shop.

Ready to shop?

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

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

Say "Cheese" for Oral Health!

From short roots to crowding of teeth patients with various types of homocystinuria experience a wide range of eye issues. While some of these dental issues are better documented, other aspects aren't.

You know the  **drill, complete the Oral Health Survey**

homocystinuria.rare-x.org

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

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

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

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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