The HCU Herald

Featuring...

HCU Hero

Eleanor from the UK

HCU NETWORK AMERICA

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

HCU HERO: ELEANOR FROM THE UK

Hi, I'm Eleanor!

I'm 23 years old and live in the UK. It wasn't until I was 10 years old that I was diagnosed with classical HCU.

As a young child and before my diagnosis, I definitely experienced symptoms but didn't have an answer as to why. I didn't speak much until I was about 3 years old and it was also around this time that my vision began to deteriorate drastically. I was always very tired, my urine smelled strange, and I was sick a lot. I didn't hit my milestones like my sister did, so it was worrisome for everyone.

In school I had difficulty focusing and completing tasks. I think maybe part of this was just that I didn't feel well nor did I have the energy to sustain focus.

I also had a really hard time seeing the board, which made things difficult for me as well. I got glasses around 8 years old and since then my vision has improved and thankfully, I haven't really had any further issues with my vision.

For over 8 years, I was misdiagnosed. It was incredibly frustrating because while I knew something was wrong, doctors kept fobbing me and my mum off and would often compare me and my sister.

Finally, one of my doctors ran a blood test, and shortly after, we got a phone call saying that we needed to come in. They took me and my mum into a room and delivered the news that I had homocystinuria. They also ran a blood test on my sister, and her test was negative. It was just me.

As I listened to the doctor deliver the news of my diagnosis, it all went over my head. I just remember feeling like a failure, and I was angry and upset. But, receiving my diagnosis was in some ways a relief that I wasn't going crazy, and a relief to my mum because she had known all along that something wasn't right. At least now we had an answer.

The only thing I really remember the doctor saying was that I couldn't eat a lot of protein anymore. I thought "What in the world is going on? I used to eat protein and now I can't?" When you're 10 years old, you don't fully realize what's going on. I didn't understand this new diagnosis and I also couldn't understand why there were now certain things I couldn't eat.

Receiving my diagnosis was in some ways a relief that I wasn't going crazy, and a relief to my mum because she had known all along that something wasn't right. At least now we had an answer.

It was very hard having a new diet because I couldn't eat so many things that I enjoyed which made me sad and angry. I used to eat a lot of chicken and all of a sudden, no more. As I've grown up I've gotten used to it and now my diet is really healthy. Some of my favorite low-protein meals are pesto pasta with veggies and low-protein cheese toast. My current treatment consists of a low-protein diet, medical formula, Cystadane, folic acid, and Vitamin D3.

I wish I had been diagnosed earlier because I wouldn't have had such trouble adjusting to a low-protein diet. My late diagnosis also resulted in a learning disability and other special needs that make my life hard. I have had support in school for education, physio, occupational therapy, speech therapy, and play therapy. I am doing really well now and with support, I have a job working in textiles. I live at home and have support with all the things I need to do. I enjoy meeting friends and having a good social life, which also includes meeting with people from disability groups. In my free time, I love couponing and weaving.

I hope that one day we will have some sort of treatment that would allow me to eat a more normal diet. I also have plans to raise money for HCU charity. Finally, I hope to one day live in my own home like my sister and maybe get married and have a family, but we'll see what the future holds!

To read Eleanor's story on our website, visit
https://hcunetworkamerica.org/eleanor-uk/

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!

Easy Lemon Garlic Pasta

***Makes 2 servings* | .5 grams protein per serving**

Ingredients:

- 124 g Aproten Linguini, cooked according to package
- 2 TBSP Boursin Dairy Free Spread
- 1 tsp Lemon Zest
- 1 TBSP Lemon Juice
- 15 g Follow Your Heart Parmesan Style Shredded
- 1 TBSP Scallions, green parts, sliced

Directions:

1. Cook pasta according to package instructions. Drain and rinse, set aside.

2. In a small sauce pan add a little olive oil or a tablespoon of butter and heat over medium heat. Add the boursin and cook until melted. Add the lemon juice and stir. Add the green onions and parmesan cheese. Cook until thickened. Add the pasta and fold to coat. Top with more cheese and parsley. Serve immediately.

November 2023, HCU & You: Ask Methia

Dear Methia,

Help me expand my low-pro menu!!

What I'm about to share with you will probably be of no surprise because I feel like I'm not alone here: FOOD IS BORING! I am so sick of eating the same things repeatedly. At this point, I feel like every day looks exactly the same. I know there are more low protein products on the market than ever before, but I don't always know where to find them. On top of that, I'm not sure I can visualize exactly how new foods will be able to fit into my diet. I am willing to cook, but I need ideas! Can you help make eating more fun?

Sincerely,
What's for Dinner?

Dear What's for Dinner?,
Eating the same thing every day can be a major snooze! There's no sugar-coating that being on a protein-restricted diet truly limits options. However, you're absolutely right in that there are more options now than there ever have been. On top of that, families and organizations have gone the extra mile in low-protein diet development and are now sharing recipes with the entire community. Let's not recreate the wheel here - I'll tell you how to make the most of what's around!

Look for recipes from dietitians, patient organizations, and non-profits dedicated to the metabolic community. These organizations are typically run by families who are also navigating a low-protein diet and have lovingly spent their time on recipe creation and modifications. Examples include the Cook For Love website and cookbooks written by author and renowned metabolic dietitian Virginia Schuett (Low Protein Cookery for PKU, Apples to Zucchini: A Collection of Favorite Low Protein Recipes). You can even find recipes on patient organization sites that predominantly serve other metabolic disorders. For example, the urea cycle disorder website UCDandYou.com provides low-protein recipes that not only calculate the amount of protein per serving, but also the amount of methionine, leucine, tyrosine, and phenylalanine for people with amino acid disorders.

Utilize apps and nutrient databases to see how new foods can fit into your diet. Everyone with homocystinuria has a different nutrition prescription. These apps help you to individualize your meal plan by calculating the amount of calories, protein/methionine, medical food, and other macro and micronutrients as you plan your daily intake. They allow you to search an expansive database that typically calculates protein more accurately than manually calculating using food labels. Examples include How Much Phe, and soon, the FLOK app (formerly PKU News, and now serving all inherited disorders of protein metabolism). Using these apps to plan meals might really shock you - in a good way - when you see how much can actually be included in your diet while still meeting your protein and calorie goals!

Connect with other patients in your clinic or online. Ask your geneticist and dietitian if there are other local patients and families who want to meet other people on low-protein diets. This is an amazing way to not only meet new friends who truly understand the challenges you're facing but to also learn from one another. You could center meet-ups around recipe exchanges, play groups or book clubs (where everyone brings a low-protein dish with the recipe), and even group cooking classes.

It's so important to continue to "make food fun," despite many restrictions. Connecting with your community can really open your eyes to new ideas. Once you implement some new products and recipes, who knows - you may be creating some low protein recipes of your own!

Sincerely,
Methia

What is Giving Tuesday?

GivingTuesday is a global generosity movement unleashing the power of radical generosity. GivingTuesday was created in 2012 as a simple idea: a day that encourages people to do good. Since then, it has grown into a year-round global movement that inspires hundreds of millions of people to give, collaborate, and celebrate generosity. This year, GivingTuesday will be **November 28**.

How do I get involved?

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

To get started, set up your own GivingTuesday fundraiser on GoFundMe.com, Facebook, or Instagram! Setting up your fundraiser on these platforms is simple and HCU Network America receives 100% of the donations!

- Start to reach out to your friends and family in advance and get them to pledge a donation first thing on November 28th!
- Let them know that by giving to HCU Network America, they are supporting programs and resources that directly benefit the patients and caregivers of our community.
- Remind them that all donations are tax-deductible AND will be matched by 3 anonymous donors (up to $25,000!)

How do I set up a GoFundMe fundraiser?

It's easy! Select 'charity' and type in HCU Network America. From there, just follow the prompts! GoFundMe is great because you can link in a video if you'd like. Consider a quick video telling how our programs have made a positive impact on you, along with your ask. Or, you can always post an image along with text.

GIVING TUESDAY

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.

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://hcunetworkamerica.org/donate
3. Text HCU2023 to 44-321
4. Scan the QR Code

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?

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.

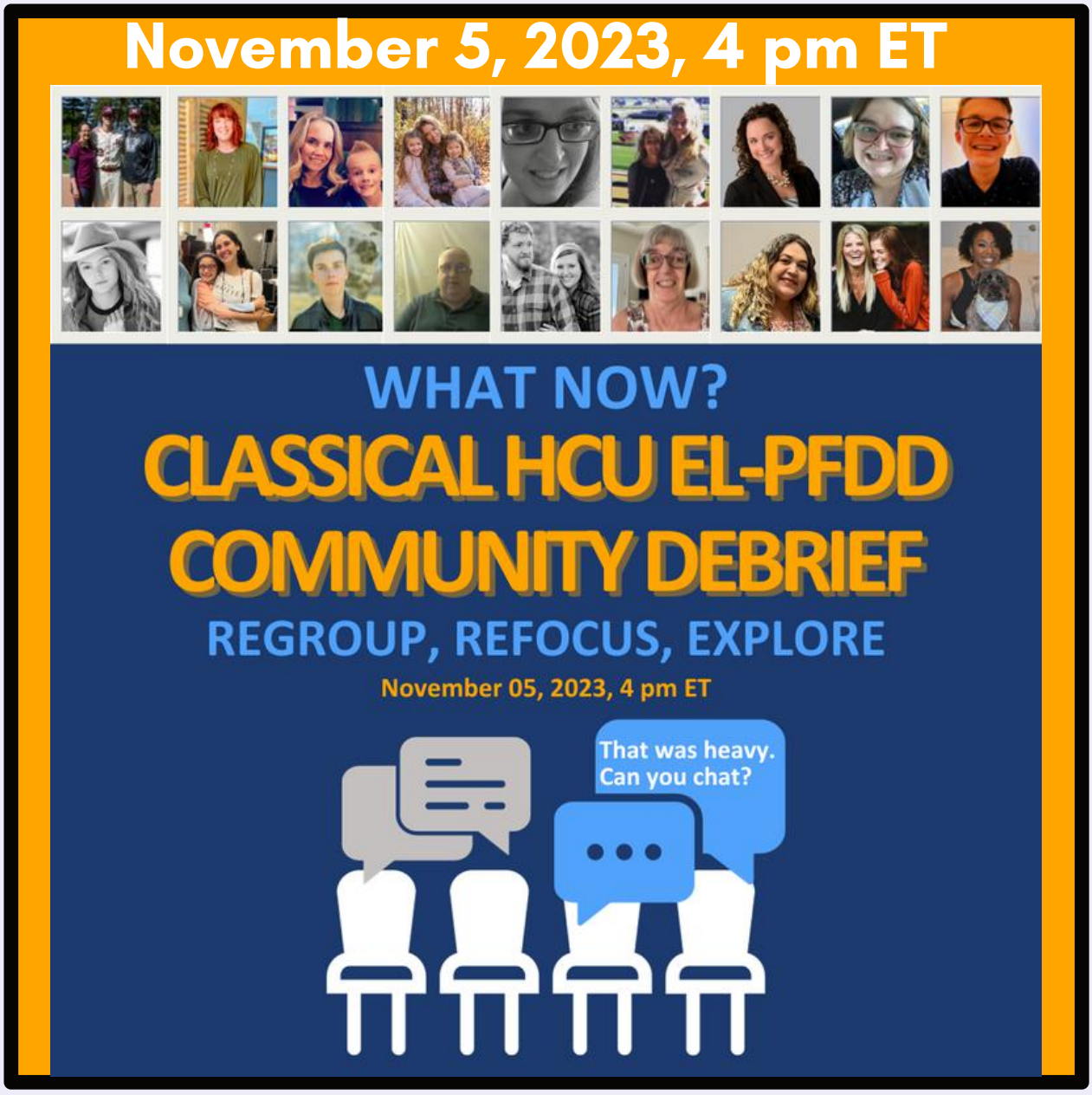
Shop normally (no special codes, no special anything) at any of about 1,800 stores. The Button is working in the background to let them know you're helping when you shop.

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!

UPCOMING EVENTS

A lot of big feelings come about as a result of EL-PFDDs. This will be an opportunity for our community to regroup, refocus, and explore the topics, corresponding feelings, coping mechanisms, and a path forward.



This is only open to patients, parents, and caregivers. Medical professionals, researchers, and industry are respectfully asked to not attend as this is a private time for the community.

Click **here** to register to attend!

UPCOMING EVENTS

CLASSICAL HCU TEEN MEETUP

Classical HCU teen (10-22) meetups are an opportunity for teens with Classical HCU to connect, learn, and share their 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 at: https://bit.ly/hcuteen

HCU AWANRESS MONTH: ADVOCATES IN ACTION!

Juana from Uruguay

Juana is 11 years old and is the only patient diagnosed with homocystinuria in Uruguay! During HCU Awareness Month, she gave a presentation on HCU to her 5th-grade class.

Great job Juana! Thank you for helping spread awareness for our community!

Kelly from Michigan

Kelly also spent HCU Awareness Month engaged in advocacy! She spoke with a healthcare system that is contracted through her local school system about what it's like to live with HCU, including some of the struggles that can impact performance in school, like poor eyesight and issues with low energy. She also highlighted some of the classroom accommodations that can help - preferential seating, access to low-protein snacks, and time to take medication or formula. She highlighted some of the issues that she experienced while in school and shared ideas of how the educational experience could be made easier for someone with HCU.

Finally, she provided them with resources where they could learn more about HCU and other Inborn Errors of Metabolism. ***Amazing work, Kelly!***

2024 Family Conference

Click here to register to attend!

Day 1
June 29, 2024

- 07:00 Registration, Breakfast and Vendors
- 08:30 Keynote Speaker: Moving Mountains
- 10:30 Panel: Aging with a Homocystinuria
- 12:00 Lunch
- 1:30 Breakout 1
- 2:45 Breakout 2
- 4:00 Breakout 3
- 6:00 Dinner and HCU Hero Award Reception

Day 2
June 30, 2024

- 07:00 Breakfast and Vendors open
- 08:30 Newborn Screening
- 09:00 Research Updates
- 11:00 HCU Data Collection Program Update
- 12:00 Ask the Experts
- 12:30 Closing Comments and Lunch

20 MOVING MOUNTAINS 24
HCU Network America
CONFERENCE

KidsZone is available for ages 3-18 | Schedule is subject to change

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

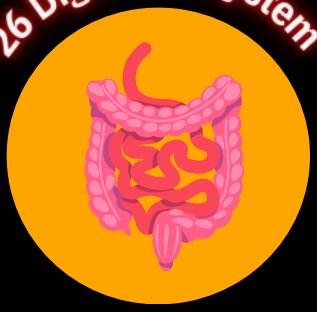
It's not Halloween, it's just HCU

Of the 51 participants who took the Health and Development Survey here are the top 5 areas the data is pointing too

32 Vision issues

27 Central Nervous System issues

26 Digestive System



25 Bone, Cartilage, and Connective Tissue issues

24 Behavior or Psychiatric conditions

Complete the Bone, Cartilage and Connective Tissue Survey: https://homocystinuria.rare-x.org/

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

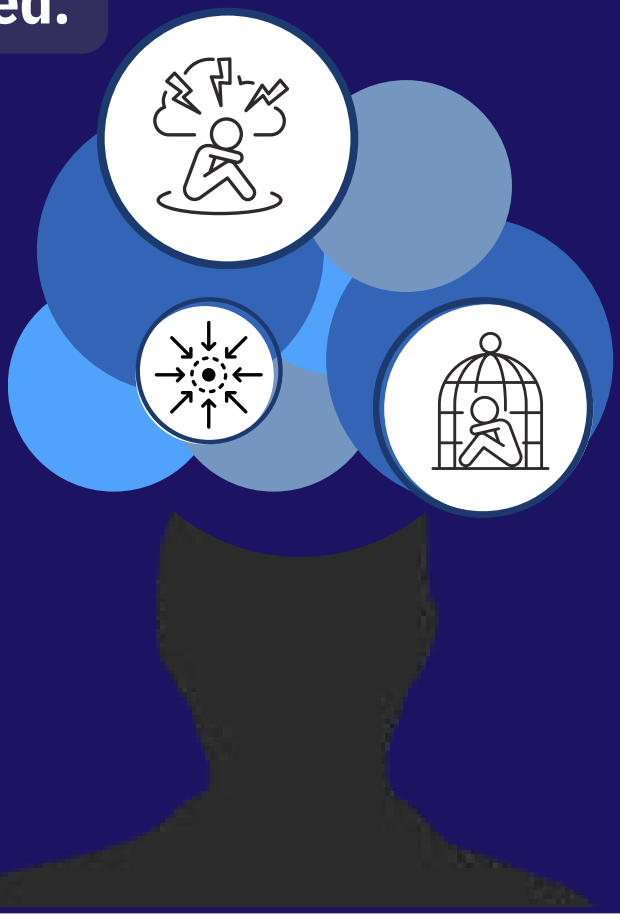
Want more to learn what other tricks and treats the HCU DCP is sharing?

Watch the video (it's only 3 minutes)

HOMOCYSTINURIAS

DATA COLLECTION PROGRAM

In a study from a 2019 publication, "Revising the psychiatric phenotype of homocystinuria", 16 of the 25 patients in the sample (64%) reported psychiatric symptoms, including a high prevalence of both anxiety (32%) and depression (32%). Deficit–hyperactivity disorder (ADHD), oppositional defiant disorder (ODD), mood swings, hallucinations, and suicidal thoughts have also been reported.



Complete the Behavior Survey

homocystinuria.rare-x.org

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

MEDICAL ASSISTANCE

What is the purpose of this program?

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

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

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

Who is eligible to apply?

This program is designed to help patients who:

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

What is the application process?

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

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

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

NORD is Here for You

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

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

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

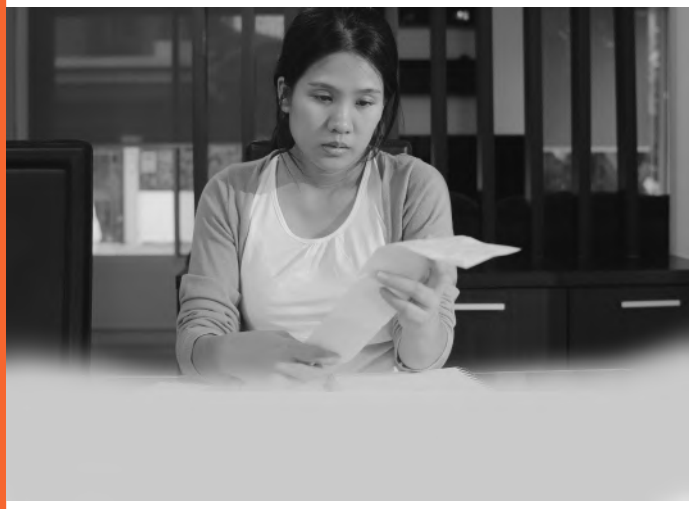
Monday-Thursday 8:30am – 7:00pm ET
Friday 8:30 am – 6:00pm ET

203-616-4327

203-635-4163

hcu@rarediseases.org

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



What assistance does NORD provide?

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

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

Awards are issued for a calendar year.

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

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

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

How does NORD demonstrate compliance with regulations required of charities?

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

NORD®

rarediseases.org

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:
https://bit.ly/3WUgYQS

MA-PE-23-0050 Sep 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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